(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property Organization
International Bureau

(43) International Publication Date
22 November 2018 (22.11.2018)

WIPO | PCT

(10) International Publication Number
WO 2018/213708 A1

(51) International Patent Classification:
C12N 15/10 (2006.01) *C12N 9/78* (2006.01)
C12N 9/22 (2006.01) *C12N 15/62* (2006.01)

(21) International Application Number:
PCT/US2018/033394

(22) International Filing Date:
18 May 2018 (18.05.2018)

(25) Filing Language: English

(26) Publication Language: English

(30) Priority Data:

62/508,293	18 May 2017 (18.05.2017)	US
62/561,663	21 September 2017 (21.09.2017)	US
62/568,133	04 October 2017 (04.10.2017)	US
62/609,957	22 December 2017 (22.12.2017)	US

(71) Applicants: **THE BROAD INSTITUTE, INC.** [US/US]; 415 Main Street, Cambridge, Massachusetts 02142 (US). **MASSACHUSETTS INSTITUTE OF TECHNOLOGY** [US/US]; 77 Massachusetts Avenue, Cambridge, Massachusetts 02139 (US). **PRESIDENT AND FELLOWS OF HARVARD COLLEGE** [US/US]; 17 Quincy Street, Cambridge, Massachusetts 02138 (US).

(72) Inventors: **ZHANG, Feng**; 100 Pacific Street, Apt. 11, Cambridge, Massachusetts 02139 (US). **COX, David**; 375A Harvard Street, Apt. 12A, Cambridge, Massachusetts 02138 (US). **GOOTENBERG, Jonathan**; 100 Memorial Drive, Apt. 15-14A, Cambridge, Massachusetts 02142 (US). **ABUDAYYEH, Omar O.**; 60 Phillips Street, Boston, Massachusetts 02114 (US). **ZETSCHE, Bernd**; 383 Washington Street, Gloucester, Massachusetts 01930 (US). **STRECKER, Jonathan**; 415 Main Street, Cambridge, Massachusetts 02142 (US).

(74) Agent: **TALAPATRA, Sunit** et al.; Foley & Lardner LLP, 3000 K Street, N.W., Suite 600, Washington, District of Columbia 20007-5143 (US).

(81) Designated States *(unless otherwise indicated, for every kind of national protection available)*: AE, AG, AL, AM, AO, AT, AU, AZ, BA, BB, BG, BH, BN, BR, BW, BY, BZ, CA, CH, CL, CN, CO, CR, CU, CZ, DE, DJ, DK, DM, DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IR, IS, JO, JP, KE, KG, KH, KN, KP,

(54) Title: SYSTEMS, METHODS, AND COMPOSITIONS FOR TARGETED NUCLEIC ACID EDITING

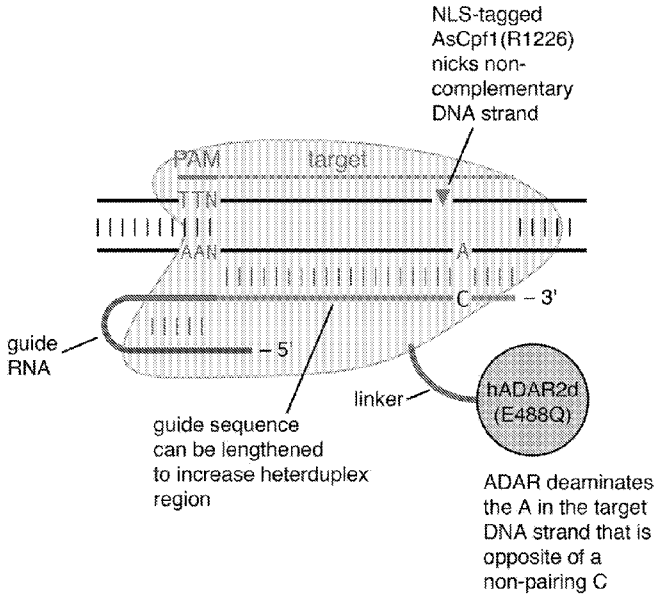


FIGURE 1

(57) Abstract: The invention provides for systems, methods, and compositions for targeting and editing nucleic acids. In particular, the invention provides non-naturally occurring or engineered DNA-targeting systems comprising a DNA-targeting Cpfl protein, at least one guide molecule, and at least one adenosine deaminase protein or catalytic domain thereof.

[Continued on next page]

WO 2018/213708 A1

KR, KW, KZ, LA, LC, LK, LR, LS, LU, LY, MA, MD, ME, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PA, PE, PG, PH, PL, PT, QA, RO, RS, RU, RW, SA, SC, SD, SE, SG, SK, SL, SM, ST, SV, SY, TH, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, ZA, ZM, ZW.

(84) Designated States *(unless otherwise indicated, for every kind of regional protection available)*: ARIPO (BW, GH, GM, KE, LR, LS, MW, MZ, NA, RW, SD, SL, ST, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, RU, TJ, TM), European (AL, AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HR, HU, IE, IS, IT, LT, LU, LV, MC, MK, MT, NL, NO, PL, PT, RO, RS, SE, SI, SK, SM, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, KM, ML, MR, NE, SN, TD, TG).

Declarations under Rule 4.17:

— *as to applicant's entitlement to apply for and be granted a patent (Rule 4.17(ii))*

Published:

— *with international search report (Art. 21(3))*

— *before the expiration of the time limit for amending the claims and to be republished in the event of receipt of amendments (Rule 48.2(h))*

SYSTEMS, METHODS, AND COMPOSITIONS FOR TARGETED NUCLEIC ACID EDITING

RELATED APPLICATIONS AND INCORPORATION BY REFERENCE

[0001] This application claims priority to U.S. Provisional Application No. 62/508,293 filed May 18, 2017, U.S. Provisional Application No. 62/561,663 filed September 21, 2017, U.S. Provisional Application No. 62/568,133 filed October 4, 2017, and U.S. Provisional Application No. 62/609,957 filed December 22, 2017, each of which is incorporated herein by reference in its entirety

STATEMENT AS TO FEDERALLY SPONSORED RESEARCH

[0002] This invention was made with government support under grant numbers MH100706 and MH110049 awarded by the National Institutes of Health. The government has certain rights in the invention.

FIELD OF THE INVENTION

[0003] The present invention generally relates to systems, methods, and compositions for targeting and editing nucleic acids, in particular for programmable deamination of adenine at a target locus of interest.

BACKGROUND

[0004] Recent advances in genome sequencing techniques and analysis methods have significantly accelerated the ability to catalog and map genetic factors associated with a diverse range of biological functions and diseases. Precise genome targeting technologies are needed to enable systematic reverse engineering of causal genetic variations by allowing selective perturbation of individual genetic elements, as well as to advance synthetic biology, biotechnological, and medical applications. Although genome-editing techniques such as designer zinc fingers, transcription activator-like effectors (TALEs), or homing meganucleases are available for producing targeted genome perturbations, there remains a need for new genome engineering technologies that employ novel strategies and molecular mechanisms and are affordable, easy to set up, scalable, and amenable to targeting multiple positions within the eukaryotic genome. This would provide a major resource for new applications in genome engineering and biotechnology.

[0005] Programmable deamination of cytosine has been reported and may be used for correction of A→G and T→C point mutations. For example, Komor et al., Nature (2016) 533:420–424 reports targeted deamination of cytosine by APOBEC1 cytidine deaminase in a non-targeted DNA stranded displaced by the binding of a Cas9-guide RNA complex to a targeted DNA strand, which results in conversion of cytosine to uracil. See also Kim et al., Nature Biotechnology (2017) 35:371-376; Shimatani et al., Nature Biotechnology (2017) doi:10.1038/nbt.3833; Zong et al., Nature Biotechnology (2017) doi:10.1038/nbt.3811; Yang Nature Communication (2016) doi:10.1038/ncomms13330.

[0006] However, A→G and T→C point mutations only represent about 12% of known pathogenic SNPs, while G→A and C→T point mutations represent about 47% of known pathogenic SNPs and are not addressable by deamination of cytosine. Novel systems and methods are required for correction of these point mutations and pathogenic SNPs.

SUMMARY OF THE INVENTION

[0007] At least a first aspect of the invention relates to a method of modifying an Adenine in a target locus of interest, the method comprising delivering to the locus: (a) a Cpf1 nickase protein; (b) a guide molecule which comprises a guide sequence linked to a direct repeat sequence; and (c) an adenosine deaminase protein or catalytic domain thereof; wherein the adenosine deaminase protein or catalytic domain thereof is covalently or non-covalently linked to the Cpf1 nickase protein or the guide molecule or is adapted to link thereto after delivery; wherein guide molecule forms a complex with the Cpf1 nickase protein and directs the complex to bind a first DNA strand at the target locus of interest, wherein the guide sequence is capable of hybridizing with a target sequence comprising the Adenine within the first DNA strand to form a heteroduplex that comprises a non-pairing Cytosine opposite to the Adenine; wherein the Cpf1 nickase protein nicks a second DNA strand at the target locus of interest displaced by the formation of the heteroduplex; and wherein the adenosine deaminase protein or catalytic domain thereof deaminates the Adenine in the heteroduplex opposite to the non-pairing Cytosine.

[0008] In some embodiments, the adenosine deaminase protein or catalytic domain thereof is fused to N- or C-terminus of the Cpf1 nickase protein. In some embodiments, the adenosine deaminase protein or catalytic domain thereof is fused to the Cpf1 nickase protein by a linker. In some embodiments, the linker is $(GGGGS)_{3-11}$ (SEQ ID NOS: 1-9), GSG_5 (SEQ ID NO: 10) or LEPGEKPYKCPECGKSFSQSGALTRHQRTHTR (SEQ ID NO: 11).

[0009] In some embodiments, the adenosine deaminase protein or catalytic domain thereof is linked to an adaptor protein and the guide molecule or the Cpf1 nickase protein comprises an aptamer sequence capable of binding to the adaptor protein. In some embodiments, the adaptor sequence is selected from MS2, PP7, Qβ, F2, GA, fr, JP501, M12, R17, BZ13, JP34, JP500, KU1, M11, MX1, TW18, VK, SP, FI, ID2, NL95, TW19, AP205, ϕCb5, ϕCb8r, ϕCb12r, ϕCb23r, 7s and PRR1.

[0010] In some embodiments, the adenosine deaminase protein or catalytic domain thereof is inserted into an internal loop of the Cpf1 nickase protein.

[0011] In some embodiments, the Cpf1 nickase protein is a nickase which nicks the non-target strand, i.e. the strand complementary to the strand comprising the target sequence and hybridizing with the guide sequence. In some embodiments, the Cpf1 nickase protein comprises a mutation in the Nuc domain. In some embodiments, the Cpf1 nickase protein comprises a mutation corresponding to R1226A in AsCpf1.

[0012] In some embodiments, the Cpf1 nickase has part or all of the Nuc domain removed. In one specific embodiment, amino acids 1076 to 1258 of AsCpf1 is removed and replaced with a linker (e.g., GSGG or GGSGGS linker).

[0013] In some embodiments, the CRISPR-Cas protein is a dead Cpf1 which comprises a mutation in the RuvC domain. In some embodiments, the CRISPR-Cas protein is a dead Cpf1 and comprises a mutation corresponding to D908A or E993A in AsCpf1. In some embodiments, the dead Cpf1 has part or all of the Nuc domain removed. In one specific embodiment, amino acids 1076 to 1258 of AsCpf1 is removed and replaced with a linker (e.g., GSGG or GGSGGS linker).

[0014] In some embodiments, the guide molecule binds to Cpf1 and is capable of forming a heteroduplex of about 24 nt with the target sequence. In some embodiments, the guide molecule binds to Cpf1 and is capable of forming a heteroduplex of more than 24 nt with the target sequence.

[0015] In some embodiments, the adenosine deaminase is a human, squid or Drosophila adenosine deaminase. In some embodiments, the adenosine deaminase has been modified to increase_activity against a DNA-RNA heteroduplex . In some embodiments, the adenosine deaminase is a mutated hADAR2d comprising the mutation E488Q or a mutated hADAR1d comprising the mutation E1008Q.

[0016] In some embodiments, the method comprises, determining the target sequence of interest and selecting the adenosine deaminase which most efficiently deaminates the Adenine present in the target sequence.

[0017] In some embodiments, the Cpf1 nickase protein is derived from a bacterial species selected from the group consisting of Francisella tularensis, Prevotella albensis, Lachnospiraceae bacterium, Butyrivibrio proteoclasticus, Peregrinibacteria bacterium, Parcubacteria bacterium, Smithella sp., Acidaminococcus sp., Lachnospiraceae bacterium, Candidatus Methanoplasma termitum, Eubacterium eligens, Moraxella bovoculi, Leptospira inadai, Porphyromonas crevioricanis, Prevotella disiens and Porphyromonas macacae, Succinivibrio dextrinosolvens, Prevotella disiens, Flavobacterium branchiophilum, Helcococcus kunzii, Eubacterium sp., Microgenomates (Roizmanbacteria) bacterium, Flavobacterium sp., Prevotella brevis, Moraxella caprae, Bacteroidetes oral, Porphyromonas cansulci, Synergistes jonesii, Prevotella bryantii, Anaerovibrio sp., Butyrivibrio fibrisolvens, Candidatus Methanomethylophilus, Butyrivibrio sp., Oribacterium sp., Pseudobutyrivibrio ruminis and Proteocatella sphenisci.

[0018] In some embodiments, the natural PAM sequence is TTN, where N is A/C/G or T and the CRISPR-Cas protein is FnCpf1 or wherein the PAM sequence is TTTV, where V is A/C or G and the CRISPR-Cas protein is PaCpf1p, LbCpf1 or AsCpf1.

[0019] In some embodimentsthe Cpf1 nickase protein has been modified and recognizes an altered PAM sequence.

[0020] In some embodiments, the target locus of interest is within a cell. In some embodiments, the cell is a eukaryotic cell. In some embodiments, the cell is a non-human animal cell. In some embodiments, the cell is a human cell. In some embodiments, the cell is a plant cell.

[0021] In some embodiments, the target locus of interest is within an animal. In some embodiments, the target locus of interest is within a plant. In some embodiments, the target locus of interest is comprised in a DNA molecule *in vitro*.

[0022] In some embodiments, the components (a), (b) and (c) are delivered to the cell as a ribonucleoprotein complex.

[0023] In some embodiments, the components (a), (b) and (c) are delivered to the cell as one or more polynucleotide molecules. In some embodiments, the one or more polynucleotide molecules comprise one or more mRNA molecules encoding components (a) and/or (c).

[0024] In some embodiments, the one or more polynucleotide molecules are comprised within one or more vectors. In some embodiments, the one or more polynucleotide molecules comprise one or more regulatory elements operably configured to express the Cpf1 nickase protein, the guide molecule, and the adenosine deaminase protein or catalytic

domain thereof, optionally wherein the one or more regulatory elements comprise inducible promoters.

[0025] In some embodiments, the Cpf1 nickase protein and optionally the adenosine deaminase protein or catalytic domain thereof comprise one or more heterologous nuclear localization signal(s) (NLS(s)).

[0026] In some embodiments, the one or more polynucleotide molecules or the ribonucleoprotein complex are delivered via particles, vesicles, or one or more viral vectors.

[0027] In some embodiments, the particles comprise a lipid, a sugar, a metal or a protein. In some embodiments, the particles comprise lipid nanoparticles.

[0028] In some embodiments, the vesicles comprise exosomes or liposomes. In some embodiments, the one or more viral vectors comprise one or more of adenovirus, one or more lentivirus or one or more adeno-associated virus.

[0029] In some embodiments, the method modifies a cell, a cell line or an organism by manipulation of one or more target sequences at genomic loci of interest.

[0030] At least a second aspect of the invention relates to a method for treating or preventing a disease using the method described herein, wherein the deamination of the Adenine at the target locus of interest remedies a disease caused by a G→A or C→T point mutation or a pathogenic SNP. In some embodiments, the disease is selected from cancer, haemophilia, beta-thalassemia, Marfan syndrome and Wiskott-Aldrich syndrome.

[0031] At least a third aspect of the invention relates to a method for knock-out or knock-down an undesirable activity of a gene or regulatory element thereof, wherein the deamination of the Adenine at the target locus of interest inactivates a target gene or a target regulatory element at the target locus.

[0032] At least a fourth aspect of the invention relates to a modified cell obtained from the method described above, or progeny thereof, wherein the cell comprises a hypoxanthine or a guanine in replace of the Adenine in the target locus of interest compared to a corresponding cell not subjected to the method.

[0033] In some embodiments, the modified cell is a eukaryotic cell. In some embodiments, the modified cell is an animal cell. In some embodiments, the modified cell is a human cell. In some embodiments, the modified cell is a plant cell.

[0034] In some embodiments, the modified cell is a therapeutic T cell. In some embodiments, the modified cell is an antibody-producing B cell.

[0035] At least a fifth aspect of the invention relates to a non-human animal or a plant comprising the modified cell described herein.

[0036] At least a sixth aspect of the invention relates to a method for cell therapy, comprising administering to a patient in need thereof the modified cell described herein, wherein the presence of the modified cell remedies a disease in the patient.

[0037] At least a seventh aspect of the invention relates to an engineered, non-naturally occurring system suitable for modifying an Adenine in a target locus of interest, comprising: a guide molecule which comprises a guide sequence linked to a direct repeat, or a nucleotide sequence encoding the guide molecule; a Cpf1 nickase protein, or one or more nucleotide sequences encoding the Cpf1 nickase protein; an adenosine deaminase protein or catalytic domain thereof, or one or more nucleotide sequences encoding; wherein the adenosine deaminase protein or catalytic domain thereof is covalently or non-covalently linked to the Cpf1 nickase protein or the guide molecule or is adapted to link thereto after delivery; wherein the guide sequence is capable of hybridizing with a target sequence on a first DNA strand comprising an Adenine within the target locus, but comprises a Cytosine at the position corresponding to the Adenine; and wherein the Cpf1 nickase protein is capable of nicking a non-target sequence in a second DNA strand complementary to said first DNA strand. Accordingly, the application provides kits comprising or consisting of the components of the AD-functionalized CRISPR system described herein.

[0038] At least an eighth aspect of the invention relates to an engineered, non-naturally occurring vector system suitable for modifying an Adenine in a target locus of interest, comprising one or more vectors comprising: a first regulatory element operably linked to one or more nucleotide sequences encoding a guide molecule which comprises a guide sequence linked to a direct repeat; a second regulatory element operably linked to a nucleotide sequence encoding a Cpf1 nickase protein; and optionally a nucleotide sequence encoding an adenosine deaminase protein or catalytic domain thereof which is under control of the first or second regulatory element or operably linked to a third regulatory element; wherein, if the nucleotide sequence encoding an adenosine deaminase protein or catalytic domain thereof is operably linked to a third regulatory element, the adenosine deaminase protein or catalytic domain thereof is adapted to link to the guide molecule or the Cpf1 nickase protein after expression; wherein the guide sequence is capable of hybridizing with a target sequence on a first DNA strand comprising an Adenine within the target locus, but comprises a Cytosine at the position corresponding to the Adenine; wherein components (a), (b) and (c) are located on the same or different vectors of the system; and wherein the Cpf1 nickase protein is capable of nicking a non-target sequence in a second DNA strand complementary to said first DNA strand. Accordingly, the application provides kits

comprising or consisting of vectors encoding of the components of the AD-functionalized CRISPR system described herein.

[0039] At least an ninth aspect of the invention relates to *in vitro*, *ex vivo* or *in vivo* host cell or cell line or progeny thereof comprising the engineered, non-naturally occurring system or vector system described herein.

[0040] In some embodiments, the host cell is a eukaryotic cell. In some embodiments, the host cell is an animal cell. In some embodiments, the host cell is a human cell. In some embodiments, the host cell is a plant cell.

BRIEF DESCRIPTION OF THE DRAWINGS

[0041] The novel features of the invention are set forth with particularity in the appended claims. A better understanding of the features and advantages of the present invention will be obtained by reference to the following detailed description that sets forth illustrative embodiments, in which the principles of the invention are utilized, and the accompanying drawings of which:

[0042] Figure 1 illustrates an example embodiment of the invention for targeted deamination of adenine at a target locus of interest.

[0043] Figure 2 shows the amino acid sequences of an example embodiment of a fusion protein of Cpf1 and adenosine deaminase (NLS-FLAG-AsCpf1-linker-hADAR2d(wt)).

[0044] Figure 3 shows the amino acid sequences of an example embodiment of a fusion protein of Cpf1 and adenosine deaminase (NLS-FLAG-AsCpf1(R1226A)-linker-hADAR2d(E488Q)).

[0045] Figure 4 shows the amino acid sequences of an example embodiment of a fusion protein of Cpf1 and adenosine deaminase (NLS-FLAG-AsCpf1(D908A)-linker-hADAR2d(E488Q)).

[0046] Figure 5 shows the amino acid sequences of an example embodiment of a fusion protein of Cpf1 and adenosine deaminase (NLS-FLAG-AsCpf1(E993A)-linker-hADAR2d(E488Q)).

[0047] Figure 6 shows SpCas9 & AsCpf1 fusions with huADAR2d. Four constructs for AsCpf1 and four Constructs for SpCas9 for A to G conversion were made. A nickase version for AsCpf1 (R1226A) and SpCas9 (N863A) was fused at the N or C-term with the deaminase domain of human ADAR2 (ADAR). Additionally, deletion constructs were generated by removing the HNH domain from SpCas9 or the Nuc domain from AsCpf1 to reduce steric hindrance for ADAR.

[0048] Figure 7 shows deletion constructs for ADAR fusions. Amino acids 1076 to 1258 of AsCpf1 were replaced with a GSGG linker, and amino acids 769 to 918 of SpCas9 were replaced with a GGSGGS linker.

[0049] Figure 8 shows expression of ADAR fusions in HEK cells. HEK293T cells were transfected with different ADAR fusion constructs or HNH/Nuc deletion constructs to confirm protein expression. Cells were harvested two days after transfection and protein was extracted using RIPA buffer. 5ul of cell lysate was used for western blot, using antibodies against a Flag (SpCas9) or HA (AsCpf1) tag.

[0050] Figure 9 shows guide design for programmed A-to-G conversion of mRNA target in HEK293 cells.

[0051] Figure 10 shows results of programmed A-to-G conversion of mRNA target in HEK293 cells.

[0052] Figure 11 shows targeted A-to-G base editing of human DNMT1 in HEK293 cells. AsCpf1(R1226A)-ADAR2d and AsCpf1(deltaNuc)-ADAR2d fusion constructs, in complex with guide RNA targeting human DNMT1 gene, each showed a detectable level of targeted A-to-G base editing, in contrast to wtAsCpf1 and ADAR2d control constructs.

[0053] The figures herein are for illustrative purposes only and are not necessarily drawn to scale.

DETAILED DESCRIPTION

[0054] Various embodiments are described hereinafter. It should be noted that the specific embodiments are not intended as an exhaustive description or as a limitation to the broader aspects discussed herein. One aspect described in conjunction with a particular embodiment is not necessarily limited to that embodiment and can be practiced with any other embodiment(s).

[0055] **Methods for Targeted Deamination of Adenine**

[0056] In one aspect the present invention provides methods for targeted deamination of adenine in a DNA, more particularly in a locus of interest. According to the methods of the invention, the adenosine deaminase (AD) protein is recruited specifically to the relevant Adenine in the locus of interest by a CRISPR-Cas complex which can specifically bind to a target sequence. In order to achieve this, the adenosine deaminase protein can either be covalently linked to the CRISPR-Cas enzyme or be provided as a separate protein, but adapted so as to ensure recruitment thereof to the CRISPR-Cas complex.

[0057] In particular embodiments, of the methods of the present invention, recruitment of the adenosine deaminase to the target locus is ensured by fusing the adenosine deaminase

or catalytic domain thereof to the CRISPR-Cas protein, which is a Cpf1 protein. Methods of generating a fusion protein from two separate proteins are known in the art and typically involve the use of spacers or linkers. The Cpf1 protein can be fused to the adenosine deaminase protein or catalytic domain thereof on either the N- or C-terminal end thereof. In particular embodiments, the CRISPR-Cas protein is a Cpf1 protein and is linked to the N-terminus of the deaminase protein or its catalytic domain.

[0058] The term "linker" as used in reference to a fusion protein refers to a molecule which joins the proteins to form a fusion protein. Generally, such molecules have no specific biological activity other than to join or to preserve some minimum distance or other spatial relationship between the proteins. However, in certain embodiments, the linker may be selected to influence some property of the linker and/or the fusion protein such as the folding, net charge, or hydrophobicity of the linker.

[0059] Suitable linkers for use in the methods of the present invention are well known to those of skill in the art and include, but are not limited to, straight or branched-chain carbon linkers, heterocyclic carbon linkers, or peptide linkers. However, as used herein the linker may also be a covalent bond (carbon-carbon bond or carbon-heteroatom bond). In particular embodiments, the linker is used to separate the CRISPR-Cas protein and the adenosine deaminase by a distance sufficient to ensure that each protein retains its required functional property. Preferred peptide linker sequences adopt a flexible extended conformation and do not exhibit a propensity for developing an ordered secondary structure. In certain embodiments, the linker can be a chemical moiety which can be monomeric, dimeric, multimeric or polymeric. Preferably, the linker comprises amino acids. Typical amino acids in flexible linkers include Gly, Asn and Ser. Accordingly, in particular embodiments, the linker comprises a combination of one or more of Gly, Asn and Ser amino acids. Other near neutral amino acids, such as Thr and Ala, also may be used in the linker sequence. Exemplary linkers are disclosed in Maratea et al. (1985), Gene 40: 39-46; Murphy et al. (1986) Proc. Nat'l. Acad. Sci. USA 83: 8258-62; U.S. Pat. No. 4,935,233; and U.S. Pat. No. 4,751,180. For example, GlySer linkers GGS, GGGS or GSG can be used. GGS, GSG, GGGS or GGGGS linkers can be used in repeats of 3 (such as $(GGS)_3$ (SEQ ID NO: 12), (GGGGS)3) or 5, 6, 7, 9 or even 12 or more, to provide suitable lengths. In particular embodiments, linkers such as $(GGGGS)_3$ (SEQ ID NO: 13) are preferably used herein. $(GGGGS)_6$ (SEQ ID NO: 14), $(GGGGS)_9$ (SEQ ID NO: 15) or $(GGGGS)_{12}$ (SEQ ID NO: 16) may preferably be used as alternatives. Other preferred alternatives are $(GGGGS)_1$ (SEQ ID NO: 17), $(GGGGS)_2$ (SEQ ID NO: 18), $(GGGGS)_4$ (SEQ ID NO: 19),

$(GGGGS)_5$ (SEQ ID NO: 20), $(GGGGS)_7$ (SEQ ID NO: 21), $(GGGGS)_8$ (SEQ ID NO: 22), $(GGGGS)_{10}$ (SEQ ID NO: 23), or $(GGGGS)_{11}$ (SEQ ID NO: 24). In yet a further embodiment, LEPGEKPYKCPECGKSFSQSGALTRHQRTHTR (SEQ ID NO: 25) is used as a linker. In yet an additional embodiment, the linker is XTEN linker. In particular embodiments, the CRISPR-cas protein is a Cpf1 protein and is linked to the deaminase protein or its catalytic domain by means of an LEPGEKPYKCPECGKSFSQSGALTRHQRTHTR (SEQ ID NO: 26) linker. In further particular embodiments, the Cpf1 protein is linked C-terminally to the N-terminus of a deaminase protein or its catalytic domain by means of an LEPGEKPYKCPECGKSFSQSGALTRHQRTHTR (SEQ ID NO: 27) linker. In addition, N- and C-terminal NLSs can also function as linker (e.g., PKKKRKVEASSPKKRKVEAS (SEQ ID NO: 28)).

[0060] In particular embodiments of the methods of the present invention, the adenosine deaminase protein or catalytic domain thereof is delivered to the cell or expressed within the cell as a separate protein, but is modified so as to be able to link to either the Cpf1 protein or the guide molecule. In particular embodiments, this is ensured by the use of orthogonal RNA-binding protein or adaptor protein / aptamer combinations that exist within the diversity of bacteriophage coat proteins. Examples of such coat proteins include but are not limited to: MS2, Qβ, F2, GA, fr, JP501, M12, R17, BZ13, JP34, JP500, KU1, M11, MX1, TW18, VK, SP, FI, ID2, NL95, TW19, AP205, ϕCb5, ϕCb8r, ϕCb12r, ϕCb23r, 7s and PRR1. Aptamers can be naturally occurring or synthetic oligonucleotides that have been engineered through repeated rounds of in vitro selection or SELEX (systematic evolution of ligands by exponential enrichment) to bind to a specific target.

[0061] In particular embodiments of the methods and systems of the present invention, the guide molecule is provided with one or more distinct RNA loop(s) or disctinct sequence(s) that can recruit an adaptor protein. A guide molecule may be extended, without colliding with the Cpf1 protein by the insertion of distinct RNA loop(s) or disctinct sequence(s) that may recruit adaptor proteins that can bind to the distinct RNA loop(s) or distinct sequence(s). Examples of modified guides and their use in recruiting effector domains to the CRISPR-Cas complex are provided in Konermann (Nature 2015, 517(7536): 583–588). In particular embodiments, the aptamer is a minimal hairpin aptamer which selectively binds dimerized MS2 bacteriophage coat proteins in mammalian cells and is introduced into the guide molecule, such as in the stemloop and/or in a tetraloop. In these embodiments, the adenosine deaminase protein is fused to MS2. The adenosine deaminase

protein is then co-delivered together with the CRISPR-Cas protein and corresponding guide RNA.

[0062] The term "AD-functionalized CRISPR system" as used here refers to a nucleic acid targeting and editing system comprising (a) a CRISPR-Cas protein, more particularly a Cpf1 protein which is catalytically inactive or a nickase; (b) a guide molecule which comprises a guide sequence; and (c) an adenosine deaminase protein or catalytic domain thereof; wherein the adenosine deaminase protein or catalytic domain thereof is covalently or non-covalently linked to the CRISPR-Cas protein or the guide molecule or is adapted to link thereto after delivery; wherein the guide sequence is substantially complementary to the target sequence but comprises a non-pairing C corresponding to the A being targeted for deamination, resulting in an A-C mismatch in a heteroduplex formed by the guide sequence and the target sequence. For application in eukaryotic cells, the CRISPR-Cas protein and/or the adenosine deaminase are preferably NLS-tagged.

[0063] In some embodiments, the components (a), (b) and (c) are delivered to the cell as a ribonucleoprotein complex. The ribonucleoprotein complex can be delivered via one or more lipid nanoparticles.

[0064] In some embodiments, the components (a), (b) and (c) are delivered to the cell as one or more RNA molecules, such as one or more guide RNAs and one or more mRNA molecules encoding the CRISPR-Cas protein, the adenosine deaminase protein, and optionally the adaptor protein. The RNA molecules can be delivered via one or more lipid nanoparticles.

[0065] In some embodiments, the components (a), (b) and (c) are delivered to the cell as one or more DNA molecules. In some embodiments, the one or more DNA molecules are comprised within one or more vectors such as viral vectors (e.g., AAV). In some embodiments, the one or more DNA molecules comprise one or more regulatory elements operably configured to express the CRISPR-Cas protein, the guide molecule, and the adenosine deaminase protein or catalytic domain thereof, optionally wherein the one or more regulatory elements comprise inducible promoters.

[0066] In some embodiments, the CRISPR-Cas protein is a Cpf1 nickase. In some embodiments, the Cpf1 nickase comprises a mutation in the Nuc domain. In some embodiments, the Cpf1 nickase is capable of nicking a non-targeted DNA strand at the target locus of interest displaced by the formation of the heteroduplex between the targeted DNA strand and the guide molecule. Details on the aspect of the CRISPR-Cas protein in the AD-functionalized CRISPR-Cas system are provided herein elsewhere.

[0067] In some embodiments, the Cpf1 nickase comprises a mutation corresponding to R1226A in AsCpf1.

[0068] In some embodiments, the CRISPR-Cas protein is a dead Cpf1. In some embodiments, the dead Cpf1 comprises a mutation in the RuvC domain. In some embodiments, the dead Cpf1 comprises a mutation corresponding to D908A or E993A in AsCpf1.

[0069] In some embodiments of the guide molecule is capable of hybridizing with a target sequence comprising the Adenine to be deaminated within a first DNA strand at the target locus to form a heteroduplex which comprises a non-pairing Cytosine opposite to said Adenine. Upon heteroduplex formation, the guide molecule forms a complex with the Cpf1 protein and directs the complex to bind said first DNA strand at the target locus of interest. Details on the aspect of the guide of the AD-functionalized CRISPR-Cas system are provided herein below.

[0070] In some embodiments, a Cpf1 guide RNA having a canonical length (e.g., about 24 nt for AsCpf1) is used to form a heteroduplex with the target DNA. In some embodiments, a Cpf1 guide molecule longer than the canonical length (e.g., >24 nt for AsCpf1) is used to form a heteroduplex with the target DNA including outside of the Cpf1-guide RNA-target DNA complex. In certain example embodiments, the guide sequence has a length of about 20-53 nt, or about 25-53 nt, or about 29-53 nt capable of forming a DNA-RNA duplex with said target sequence. In certain other example embodiments, the guide sequence has a length of about 40-50 nt capable of forming a DNA-RNA duplex duplex with said target sequence. In certain example embodiments, the distance between said non-pairing C and the 5' end of said guide sequence is 20-30 nucleotides. In certain example embodiments, the distance between said non-pairing C and the 3' end of said guide sequence is 20-30 nucleotides. In particular embodiments, the guide sequence comprises more than one mismatch corresponding to different adenosine sites in the target DNA sequence or wherein two guide molecules are used, each comprising a mismatch corresponding to a different adenosine sites in the target RNA sequence.

[0071] In at least a first design, the AD-functionalized CRISPR system comprises (a) an adenosine deaminase fused or linked to a CRISPR-Cas protein, wherein the CRISPR-Cas protein is catalytically inactive or a nickase, and (b) a guide molecule comprising a guide sequence designed to introduce an A-C mismatch in a heteroduplex formed between the guide sequence and the target sequence. In some embodiments, the CRISPR-Cas protein and/or the adenosine deaminase are NLS-tagged, on either the N- or C-terminus or both.

[0072] In at least a second design, the AD-functionalized CRISPR system comprises (a) a CRISPR-Cas protein that is catalytically inactive or a nickase, (b) a guide molecule comprising a guide sequence designed to introduce an A-C mismatch in a heteroduplex formed between the guide sequence and the target sequence, and an aptamer sequence (e.g., MS2 RNA motif or PP7 RNA motif) capable of binding to an adaptor protein (e.g., MS2 coating protein or PP7 coat protein), and (c) an adenosine deaminase fused or linked to an adaptor protein, wherein the binding of the aptamer and the adaptor protein recruits the adenosine deaminase to the heteroduplex formed between the guide sequence and the target sequence for targeted deamination at the A of the A-C mismatch. In some embodiments, the adaptor protein and/or the adenosine deaminase are NLS-tagged, on either the N- or C-terminus or both. The CRISPR-Cas protein can also be NLS-tagged.

[0073] The use of different aptamers and corresponding adaptor proteins also allows orthogonal gene editing to be implemented. In one example in which adenosine deaminase are used in combination with cytidine deaminase for orthogonal gene editing/deamination, sgRNA targeting different loci are modified with distinct RNA loops in order to recruit MS2-adenosine deaminase and PP7-cytidine deaminase (or PP7-adenosine deaminase and MS2-cytidine deaminase), respectively, resulting in orthogonal deamination of A or C at the target loci of interested, respectively. PP7 is the RNA-binding coat protein of the bacteriophage Pseudomonas. Like MS2, it binds a specific RNA sequence and secondary structure. The PP7 RNA-recognition motif is distinct from that of MS2. Consequently, PP7 and MS2 can be multiplexed to mediate distinct effects at different genomic loci simultaneously. For example, an sgRNA targeting locus A can be modified with MS2 loops, recruiting MS2-adenosine deaminase, while another sgRNA targeting locus B can be modified with PP7 loops, recruiting PP7-cytidine deaminase. In the same cell, orthogonal, locus-specific modifications are thus realized. This principle can be extended to incorporate other orthogonal RNA-binding proteins.

[0074] In at least a third design, the AD-functionalized CRISPR system comprises (a) an adenosine deaminase inserted into an internal loop or unstructured region of a CRISPR-Cas protein, wherein the CRISPR-Cas protein is catalytically inactive or a nickase, and (b) a guide molecule comprising a guide sequence designed to introduce an A-C mismatch in a heteroduplex formed between the guide sequence and the target sequence.

[0075] CRISPR-Cas protein split sites that are suitable for inseration of adenosine deaminase can be identified with the help of a crystal structure. One can use the crystal

structure of an ortholog if a relatively high degree of homology exists between the ortholog and the intended CRISPR-Cas protein.

[0076] The split position may be located within a region or loop. Preferably, the split position occurs where an interruption of the amino acid sequence does not result in the partial or full destruction of a structural feature (e.g. alpha-helixes or β-sheets). Unstructured regions (regions that did not show up in the crystal structure because these regions are not structured enough to be "frozen" in a crystal) are often preferred options. The positions within the unstructured regions or outside loops may not need to be exactly the numbers provided above, but may vary by, for example 1, 2, 3, 4, 5, 6, 7, 8, 9, or even 10 amino acids either side of the position given above, depending on the size of the loop, so long as the split position still falls within an unstructured region of outside loop.

[0077] For Cpf1, several small stretches of unstructured regions have been predicted within the Cpf1 primary structure (see WO2016205711, the content of which is hereby incorporated by reference). Unstructured regions, which are exposed to the solvent and not conserved within different Cpf1 orthologs, are preferred sides for splits.

[0078] The following table presents non-limiting potential split regions within AsCpf1 and LbCpf1. A split site within such a region may be opportune.

Split region	AsCpf1	LbCpf1
1	575-588	566-571
2	631-645	754-757
3	653-664	-
4	818-844	-

[0079] For Fn, As and Lb Cpf1 mutants, it should be readily apparent what the corresponding position for a potential split site is, for example, based on a sequence alignment. For non-Fn, As and Lb enzymes one can use the crystal structure of an ortholog if a relatively high degree of homology exists between the ortholog and the intended Cpf1, or one can use computational prediction.

[0080] The AD-functionalized CRISPR system described herein can be used to target a specific Adenine within a DNA sequence for deamination. For example, the guide molecule can form a complex with the CRISPR-Cas protein and directs the complex to bind a target sequence at the target locus of interest. Because the guide sequence is designed to have a non-pairing C, the heteroduplex formed between the guide sequence and the target sequence comprises an A-C mismatch, which directs the adenosine deaminase to contact

and deaminate the A opposite to the non-pairing C, converting it to a Inosine (I). Since Inosine (I) base pairs with C and functions like G in cellular process, the targeted deamination of A described herein are useful for correction of undesirable G-A and C-T mutations, as well as for obtaining desirable A-G and T-C mutations.

[0081] In some embodiments, the AD-functionalized CRISPR system is used for targeted deamination in a DNA molecule *in vitro*. In some embodiments, the AD-functionalized CRISPR system is used for targeted deamination in a DNA molecule within a cell. The cell can be a eukaryotic cell, such as a animal cell, a mammalian cell, a human, or a plant cell.

[0082] The invention also relates to a method for treating or preventing a disease by the targeted deamination using the AD-functionalized CRISPR system, wherein the deamination of the A restores a healthy genotype at the target locus of interest, which remedies a disease caused by a G→A or C→T point mutation or a pathogenic SNP. Examples of disease that can be treated or prevented with the present invention include cancer, haemophilia, beta-thalassemia, Marfan syndrome, and Wiskott-Aldrich syndrome.

[0083] The invention also relates to a method for knocking-out or knocking-down an undesirable activity of a gene or regulatory element thereof, wherein the deamination of the A at the target locus of interest inactivates a target gene or a target regulatory element at the target locus. For example, in one embodiment, the targeted deamination by the AD-functionalized CRISPR system can cause a nonsense mutation resulting in a premature stop codon in an endogenous gene. This may alter the expression of the endogenous gene and can lead to a desirable trait in the edited cell. In another embodiment, the targeted deamination by the AD-functionalized CRISPR system can cause a nonconservative missense mutation resulting in a code for a different amino acid residue in an endogenous gene. This may alter the function of the endogenous gene expressed and can also lead to a desirable trait in the edited cell.

[0084] The invention also relates to a modified cell obtained by the targeted deamination using the AD-functionalized CRISPR system, or progeny thereof, wherein the modified cell comprises an I or G in replace of the A in the target locus of interest compared to a corresponding cell before the targeted deamination. The modified cell can be a eukaryotic cell, such as an animal cell, a plant cell, an mammalian cell, or a human cell.

[0085] In some embodiments, the modified cell is a therapeutic T cell, such as a T cell sutiable for CAR-T therapies. The modification may result in one or more desirable traits in the therapeutic T cell, including but not limited to, reduced expression of an immune

checkpoint receptor (e.g., PDA, CTLA4), reduced expression of HLA proteins (e.g., B2M, HLA-A), and reduced expression of an endogenous TCR.

[0086] In some embodiments, the modified cell is an antibody-producing B cell. The modification may results in one or more desirable traits in the B cell, including but not limited to, enhanced antibody production.

[0087] The invention also relates to a modified non-human animal or a modified plant. The modified non-human animal can be a farm animal. The modified plant can be an agricultural crop.

[0088] The invention further relates to a method for cell therapy, comprising administering to a patient in need thereof the modified cell described herein, wherein the presence of the modified cell remedies a disease in the patient. In one embodiment, the modified cell for cell therapy is a CAR-T cell capable of recognizing and/or attacking a tumor cell. In another embodiment, the modified cell for cell therapy is a stem cell, such as a neural stem cell, a mesenchymal stem cell, a hematopoietic stem cell, or an iPSC cell.

[0089] The invention additionally relates to an engineered, non-naturally occurring system suitable for modifying an Adenine in a target locus of interest, comprising: a guide molecule which comprises a guide sequence, or a nucleotide sequence encoding the guide molecule; a CRISPR-Cas protein, or one or more nucleotide sequences encoding the CRISPR-Cas protein; an adenosine deaminase protein or catalytic domain thereof, or one or more nucleotide sequences encoding; wherein the adenosine deaminase protein or catalytic domain thereof is covalently or non-covalently linked to the CRISPR-Cas protein or the guide molecule or is adapted to link thereto after delivery; wherein the guide sequence is capable of hybridizing with a target sequence comprising an Adenine within the target locus, but comprises a Cytosine at the position corresponding to the Adenine.

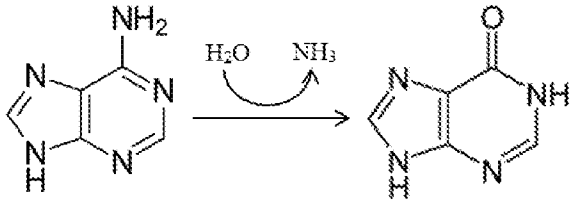
[0090] The invention additionally relates to an engineered, non-naturally occurring vector system suitable for modifying an Adenine in a target locus of interest, comprising one or more vectors comprising: a first regulatory element operably linked to one or more nucleotide sequences encoding a guide molecule which comprises a guide sequence; a second regulatory element operably linked to a nucleotide sequence encoding a CRISPR-Cas protein; and optionally a nucleotide sequence encoding an adenosine deaminase protein or catalytic domain thereof which is under control of the first or second regulatory element or operably linked to a third regulatory element; wherein, if the nucleotide sequence encoding an adenosine deaminase protein or catalytic domain thereof is operably linked to a third regulatory element, the adenosine deaminase protein or catalytic domain thereof is

adapted to link to the guide molecule or the Crispr-Cas protein after expression; wherein the guide sequence is capable of hybridizing with a target sequence comprising an Adenine within the target locus, but comprises a Cytosine at the position corresponding to the Adenine; wherein components (a), (b) and (c) are located on the same or different vectors of the system.

[0091] The invention additionally relates to in vitro, ex vivo or in vivo host cell or cell line or progeny thereof comprising the engineered, non-naturally occurring system or vector system described herein. The host cell can be a eukaryotic cell, such as an animal cell, a plant cell, an mammalian cell, or a human cell.

[0092] **Adenosine Deaminase**

[0093] The term "adenosine deaminase" or "adenosine deaminase protein" as used herein refers to a protein, a polypeptide, or one or more functional domain(s) of a protein or a polypeptide that is capable of catalyzing a hydrolytic deamination reaction that converts an adenine (or an adenine moiety of a molecule) to a hypoxanthine (or a hypoxanthine moiety of a molecule), as shown below. In some embodiments, the adenine-containing molecule is an adenosine (A), and the hypoxanthine-containing molecule is an inosine (I). The adenine-containing molecule can be deoxyribonucleic acid (DNA) or ribonucleic acid (RNA).



Adenine Hypoxanthine

[0094] According to the present disclosure, adenosine deaminases that can be used in connection with the present disclosure include, but are not limited to, members of the enzyme family known as adenosine deaminases that act on RNA (ADARs), members of the enzyme family known as adenosine deaminases that act on tRNA (ADATs), and other adenosine deaminase domain-containing (ADAD) family members. According to the present disclosure, the adenosine deaminase is capable of targeting adenine in a RNA/DNA heteroduplex. Indeed, Zheng et al. (Nucleic Acids Res. 2017, 45(6): 3369-3377) has demonstrated that ADARs can cary out adenosine to inosine editing reactions on RNA/DNA heteroduplexes. In particular embodiments, the adenosine deaminase has been

modified to increase its ability to edit DNA in a RNA/DNA heteroduplex as detailed herein below.

[0095] In some embodiments, the adenosine deaminase is derived from one or more metazoa species, including but not limited to, mammals, birds, frogs, squids, fish, flies and worms. In some embodiments, the adenosine deaminase is a human, squid or Drosophila adenosine deaminase.

[0096] In some embodiments, the adenosine deaminase is a human ADAR, including hADAR1, hADAR2, hADAR3. In some embodiments, the adenosine deaminase is a *Caenorhabditis elegans ADAR* protein, including ADR-1 and ADR-2. In some embodiments, the adenosine deaminase is a *Drosophila* ADAR protein, including dAdar. In some embodiments, the adenosine deaminase is a squid *Loligo pealeii* ADAR protein, including sqADAR2a and sqADAR2b. In some embodiments, the adenosine deaminase is a human ADAT protein. In some embodiments, the adenosine deaminase is a *Drosophila* ADAT protein. In some embodiments, the adenosine deaminase is a human ADAD protein, including TENR (hADAD1) and TENRL (hADAD2).

[0097] In some embodiments, the adenosine deaminase is a TadA protein such as E. coli TadA. See Kim et al., Biochemistry 45:6407-6416 (2006); Wolf et al., EMBO J. 21:3841-3851 (2002). In some embodiments, the adenosine deaminase is mouse ADA. See Grunebaum et al., Curr. Opin. Allergy Clin. Immunol. 13:630-638 (2013). In some embodiments, the adenosine deaminase is human ADAT2. See Fukui et al., J. Nucleic Acids 2010:260512 (2010).

[0098] In some embodiments, the adenosine deaminase is selected from:

ADAR Name	**Protein Sequence**
Homo sapiens_ADAR2_E_Q_Mutant (SEQ ID NO :29)	QLHLPQVLADAVSRLVLGKFGDLTDNFSSPHARRKVLAGVVMT TGTDVKDAKVISVSTGTKCINGEYMSDRGLALNDCHAEIISRRSL LRFLYTQLELYLNNKDDQKRSIFQKSERGGFRLKENVQFHLYIST SPCGDARIFSPHEPILEEPADRHPNRKARGQLRTKIESGQGTIPVR SNASIQTWDGVLQGERLLTMSCSDKIARWNVVGIQGSLLSIFVEP IYFSSIILGSLYHGDHLSRAMYQRISNIEDLPPLYTLNKPLLSGISN AEARQPGKAPNFSVNWTVGDSAIEVINATTGKDELGRASRLCKH ALYCRWMRVHGKVPSHLLRSKITKPNVYHESKLAAKEYQAAKA RLFTAFIKAGLGAWVEKPTEQDQFSLT*
Homo sapiens_ADAR1_E_Q_Mutant (SEQ ID NO :30)	SLGTGNRCVKGDSLSLKGETVNDCHAEIISRRGFIRFLYSELMKY NSQTAKDSIFEPAKGGEKLQIKKTVSFHLYISTAPCGDGALFDKS CSDRAMESTESRHYPVFENPKQGKLRTKVENGQGTIPVESSDIVP TWDGIRLGERLRTMSCSDKILRWNVLGLQGALLTHFLQPIYLKS

	VTLGYLFSQGHLTRAICCRVTRDGSAFEDGLRHPFIVNHPKVGR VSIYDSKRQSGKTKETSVNWCLADGYDLEILDGTRGTVDGPRNE LSRVSKKNIFLLFKKLCSFRYRRDLLRLSYGEAKKAARDYETAK NYFKKGLKDMGYGNWISKPQEEKNF
Octopus vulgaris_ADAR1_E_Q_Mutant (SEQ ID NO :31)	SVGTGNRCLTGDHLSLEGNSVNDSHAEMITRRGFLRYLYRHLLE YDAEVPNDLFEKGERSICRIKTNITFHLYISTAPCGDGALFSPRDT DSSNAKMEEENKHIHNPTFSSSVQGLLRTKVEGGQGTIPIDADFT EQTWDGIQRGERLRTMSCSDKICRWNVVGLQGALLSHFIEPIYL DSLTLGYLYDHGHLARAVCCRIERGEASVNQLLPEGYRLNHPW LGRVTACDPPRETQKTKSLSINWCYDDEKSEVLDGTAGICYTAIE KNLFSRLTKHNLYEEFKRVCRKFDRNDLLTAPSYNKAKMMATP FQTAKNVMLKKLKENNCGTWVSKPIEEEMF
Sepia_ADAR1_E_Q_Mutant (SEQ ID NO :32)	SVGTGNRCLTGDRLSLEGNSVNDSHAEMVTRRGFLRYLYKHLL EYDPEKPHDLFEKGERSLCRIKTNITFHLYISTAPCGDGALFSPRD TDSSNVKVDEENKHVHNPTFSSSVQGLLRTKVEGGQGTIPIDADF TEQTWDGIQRGERLRTMSCSDKICRWNVVGLQGALLSHFVEPIY LESLTLGYLYDHGHLARAVCCRIERGEASVNQLLPEGYRLNHPW LGRVTACDPPRETQKTKSLSINWCYDDEKSEVLDGTAGICYTAIE KNLFSRLTKHSLYEEFKKVCQKFEREDLLNVTSYNKAKMMAIPF QTAKNVMLKKLKENNCGTWVSKPIEEEMF
Octopus vulgaris_ADAR2_E_Q_Mutant (SEQ ID NO :33)	GIGTGTKCINGEHMSDRGFGVNDCHAEIIARRCFLRYIYDQLELH LSDNSDVRNSSIFELRDKGGYQLKENIQFHLYISTAPCGDARIFSP HGQDVETGDRHPNRKARGQLRTKIESGQGTIPVRTSGVIQTWDG VLEGERLLTMSCSDKIARWNVLGIQGSLLSHFMNPIYLESIILGSL YHSDHLSRAMYSRISIIENLPEPFHLNRPFLSGISSPESRQPGKAPN FGINWRKEDETFEVINAMTGRVEGGSVSRICKQALFGRFMSLYG KLSSLTGQSVTTRPTHYSDAKAAVMEYQLAKQCVFQAFQKAGL GNWVQKPIEQDQF
Sepia_ADAR2_E_Q_Mutant (SEQ ID NO :34)	GIGTGTKCINGEYMNDRGFAVNDCHAEIIARRCFLRFIYDQLEMH LSEDPEVRGQSVFELRDGGGYKLKPNIHFHLYISTAPCGDARIFSP HGQDVETGDRHPNRKARGQLRTKIESGQGTIPVRSSGFIQTWDG VLEGERLLTMSCSDKIARWNVLGIQGALLCHFMHPIYLESIILGSL YHSDHLSRAVYCRIASIENLPDLFQLNRPFLSGISSPESRQPGKAP NFGINWRRNDDTFEVINAMTGRVEGGNMSRICKQALFDRFMNL YGRLSSLTGQSVTTRPTLYSEAKAAVMEYQLAKQCVFQAFQKA GLGNWVQKPIEQDQF
Doryteusthis opalescens_ADAR2_E_Q_Mutant (SEQ ID NO :35)	GIGTGTKCINGEYMNDRGFAVNDCHAEIIARRCFLRFIYDQLELH LSDNAEVRGQSIFELRDAGGYKLKPNIQFHLYISTAPCGDARIFSP HGQDVETGDRHPNRKARGQLRTKIESGQGTIPVRSSGFIQTWDG VLEGERLLTMSCSDKIARWNVLGVQGALLCHFMHPIYLESIILGS LYHSDHLSRAVYCRIAAIENLPDLFRLNRPFLSGISSPESRQPGKA PNFGINWRRNDDSFEVINAMTGRVEGGSMSRICKQALFDRFMNL

	YGKLSSLTGQSVTTRPALYSEAKATVMEYQLAKQCVFQAFQKA GLGNWVQKPIEQDQF

[0099] In some embodiments, the adenosine deaminase protein recognizes and converts one or more target adenosine residue(s) in a double-stranded nucleic acid substrate into inosine residues (s). In some embodiments, the double-stranded nucleic acid substrate is a RNA-DNA hybrid duplex. In some embodiments, the adenosine deaminase protein recognizes a binding window on the double-stranded substrate. In some embodiments, the binding window contains at least one target adenosine residue(s). In some embodiments, the binding window is in the range of about 3 bp to about 100 bp. In some embodiments, the binding window is in the range of about 5 bp to about 50 bp. In some embodiments, the binding window is in the range of about 10 bp to about 30 bp. In some embodiments, the binding window is about 1 bp, 2 bp, 3 bp, 5 bp, 7 bp, 10 bp, 15 bp, 20 bp, 25 bp, 30 bp, 40 bp, 45 bp, 50 bp, 55 bp, 60 bp, 65 bp, 70 bp, 75 bp, 80 bp, 85 bp, 90 bp, 95 bp, or 100 bp.

[00100] In some embodiments, the adenosine deaminase protein comprises one or more deaminase domains. Not intended to be bound by theory, it is contemplated that the deaminase domain functions to recognize and convert one or more target adenosine (A) residue(s) contained in a double-stranded nucleic acid substrate into inosine (I) residues (s). In some embodiments, the deaminase domain comprises an active center. In some embodiments, the active center comprises a zinc ion. In some embodiments, during the A-to-I editing process, base pairing at the target adenosine residue is disrupted, and the target adenosine residue is "flipped" out of the double helix to become accessible by the adenosine deaminase. In some embodiments, amino acid residues in or near the active center interact with one or more nucleotide(s) 5' to a target adenosine residue. In some embodiments, amino acid residues in or near the active center interact with one or more nucleotide(s) 3' to a target adenosine residue. In some embodiments, amino acid residues in or near the active center further interact with the nucleotide complementary to the target adenosine residue on the opposite strand. In some embodiments, the amino acid residues form hydrogen bonds with the 2' hydroxyl group of the nucleotides.

[00101] In some embodiments, the adenosine deaminase comprises human ADAR2 full protein (hADAR2) or the deaminase domain thereof (hADAR2-D). In some embodiments, the adenosine deaminase is an ADAR family member that is homologous to hADAR2 or hADAR2-D.

[00102] Particularly, in some embodiments, the homologous ADAR protein is human ADAR1 (hADAR1) or the deaminase domain thereof (hADAR1-D). In some embodiments, glycine 1007 of hADAR1-D corresponds to glycine ^{487} hADAR2-D, and glutamic Acid ^{1008} of hADAR1-D corresponds to glutamic acid ^{488} of hADAR2-D.

[00103] In some embodiments, the adenosine deaminase comprises the wild-type amino acid sequence of hADAR2-D (see Fig. 2). In some embodiments, the adenosine deaminase comprises one or more mutations in the hADAR2-D sequence, such that the editing efficiency, and/or substrate editing preference of hADAR2-D is changed according to specific needs.

[00104] Certain mutations of hADAR1 and hADAR2 proteins have been described in Kuttan *et al.*, Proc Natl Acad Sci U S A. (2012) 109(48):E3295-304; Want *et al.* ACS Chem Biol. (2015) 10(11):2512-9; and Zheng *et al.* Nucleic Acids Res. (2017) 45(6):3369-337, each of which is incorporated herein by reference in its entirety.

[00105] In some embodiments, the adenosine deaminase comprises a mutation at glycine^{336} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the glycine residue at position 336 is replaced by an aspartic acid residue (G336D).

[00106] In some embodiments, the adenosine deaminase comprises a mutation at Glycine^{487} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the glycine residue at position 487 is replaced by a non-polar amino acid residue with relatively small side chains. For example, in some embodiments, the glycine residue at position 487 is replaced by an alanine residue (G487A). In some embodiments, the glycine residue at position 487 is replaced by a valine residue (G487V). In some embodiments, the glycine residue at position 487 is replaced by an amino acid residue with relatively large side chains. In some embodiments, the glycine residue at position 487 is replaced by a arginine residue (G487R). In some embodiments, the glycine residue at position 487 is replaced by a lysine residue (G487K). In some embodiments, the glycine residue at position 487 is replaced by a tryptophan residue (G487W). In some embodiments, the glycine residue at position 487 is replaced by a tyrosine residue (G487Y).

[00107] In some embodiments, the adenosine deaminase comprises a mutation at glutamic acid^{488} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the glutamic acid residue at position 488 is replaced by a glutamine residue (E488Q). In some embodiments, the glutamic acid

residue at position 488 is replaced by a histidine residue (E488H). In some embodiments, the glutamic acid residue at position 488 is replace by an arginine residue (E488R). In some embodiments, the glutamic acid residue at position 488 is replace by a lysine residue (E488K). In some embodiments, the glutamic acid residue at position 488 is replace by an asparagine residue (E488N). In some embodiments, the glutamic acid residue at position 488 is replace by an alanine residue (E488A). In some embodiments, the glutamic acid residue at position 488 is replace by a Methionine residue (E488M). In some embodiments, the glutamic acid residue at position 488 is replace by a serine residue (E488S). In some embodiments, the glutamic acid residue at position 488 is replace by a phenylalanine residue (E488F). In some embodiments, the glutamic acid residue at position 488 is replace by a lysine residue (E488L). In some embodiments, the glutamic acid residue at position 488 is replace by a tryptophan residue (E488W).

[00108] In some embodiments, the adenosine deaminase comprises a mutation at threonine^{490} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the threonine residue at position 490 is replaced by a cysteine residue (T490C). . In some embodiments, the threonine residue at position 490 is replaced by a serine residue (T490S). In some embodiments, the threonine residue at position 490 is replaced by an alanine residue (T490A). In some embodiments, the threonine residue at position 490 is replaced by a phenylalanine residue (T490F). In some embodiments, the threonine residue at position 490 is replaced by a tyrosine residue (T490Y). In some embodiments, the threonine residue at position 490 is replaced by a serine residue (T490R). In some embodiments, the threonine residue at position 490 is replaced by an alanine residue (T490K). In some embodiments, the threonine residue at position 490 is replaced by a phenylalanine residue (T490P). In some embodiments, the threonine residue at position 490 is replaced by a tyrosine residue (T490E).

[00109] In some embodiments, the adenosine deaminase comprises a mutation at valine^{493} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the valine residue at position 493 is replaced by an alanine residue (V493A). In some embodiments, the valine residue at position 493 is replaced by a serine residue (V493S). In some embodiments, the valine residue at position 493 is replaced by a threonine residue (V493T). In some embodiments, the valine residue at position 493 is replaced by an arginine residue (V493R). In some embodiments, the valine residue at position 493 is replaced by an aspartic acid residue (V493D). In some embodiments, the valine residue at position 493 is replaced by a proline

residue (V493P). In some embodiments, the valine residue at position 493 is replaced by a glycine residue (V493G).

[00110] In some embodiments, the adenosine deaminase comprises a mutation at alanine^{589} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the alanine residue at position 589 is replaced by a valine residue (A589V).

[00111] In some embodiments, the adenosine deaminase comprises a mutation at asparagine^{597} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the asparagine residue at position 597 is replaced by a lysine residue (N597K). In some embodiments, the adenosine deaminase comprises a mutation at position 597 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 597 is replaced by an arginine residue (N597R). In some embodiments, the adenosine deaminase comprises a mutation at position 597 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 597 is replaced by an alanine residue (N597A). In some embodiments, the adenosine deaminase comprises a mutation at position 597 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 597 is replaced by a glutamic acid residue (N597E). In some embodiments, the adenosine deaminase comprises a mutation at position 597 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 597 is replaced by a histidine residue (N597H). In some embodiments, the adenosine deaminase comprises a mutation at position 597 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 597 is replaced by a glycine residue (N597G). In some embodiments, the adenosine deaminase comprises a mutation at position 597 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 597 is replaced by a tyrosine residue (N597Y). In some embodiments, the asparagine residue at position 597 is replaced by a phenylalanine residue (N597F). In some embodiments, the adenosine deaminase comprises mutation N597I. In some embodiments, the adenosine deaminase comprises mutation N597L. In some embodiments, the adenosine deaminase comprises mutation N597V. In some embodiments, the adenosine deaminase comprises mutation N597M. In some embodiments, the adenosine deaminase comprises mutation N597C. In

some embodiments, the adenosine deaminase comprises mutation N597P. In some embodiments, the adenosine deaminase comprises mutation N597T. In some embodiments, the adenosine deaminase comprises mutation N597S. In some embodiments, the adenosine deaminase comprises mutation N597W. In some embodiments, the adenosine deaminase comprises mutation N597Q. In some embodiments, the adenosine deaminase comprises mutation N597D. In certain example embodiments, the mutations at N597 described above are further made in the context of an E488Q background.

[00112] In some embodiments, the adenosine deaminase comprises a mutation at serine^{599} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the serine residue at position 599 is replaced by a threonine residue (S599T).

[00113] In some embodiments, the adenosine deaminase comprises a mutation at asparagine^{613} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the asparagine residue at position 613 is replaced by a lysine residue (N613K). In some embodiments, the adenosine deaminase comprises a mutation at position 613 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 613 is replaced by an arginine residue (N613R). In some embodiments, the adenosine deaminase comprises a mutation at position 613 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 613 is replaced by an alanine residue (N613A) In some embodiments, the adenosine deaminase comprises a mutation at position 613 of the amino acid sequence, which has an asparagine residue in the wild type sequence. In some embodiments, the asparagine residue at position 613 is replaced by a glutamic acid residue (N613E). In some embodiments, the adenosine deaminase comprises mutation N613I. In some embodiments, the adenosine deaminase comprises mutation N613L. In some embodiments, the adenosine deaminase comprises mutation N613V. In some embodiments, the adenosine deaminase comprises mutation N613F. In some embodiments, the adenosine deaminase comprises mutation N613M. In some embodiments, the adenosine deaminase comprises mutation N613C. In some embodiments, the adenosine deaminase comprises mutation N613G. In some embodiments, the adenosine deaminase comprises mutation N613P. In some embodiments, the adenosine deaminase comprises mutation N613T. In some embodiments, the adenosine deaminase comprises mutation N613S. In some embodiments, the adenosine deaminase comprises mutation N613Y. In some embodiments, the adenosine deaminase

comprises mutation N613W. In some embodiments, the adenosine deaminase comprises mutation N613Q. In some embodiments, the adenosine deaminase comprises mutation N613H. In some embodiments, the adenosine deaminase comprises mutation N613D. In some embodiments, the mutations at N613 described above are further made in combination with a E488Q mutation.

[00114] In some embodiments, to improve editing efficiency, the adenosine deaminase may comprise one or more of the mutations: G336D, G487A, G487V, E488Q, E488H, E488R, E488N, E488A, E488S, E488M, T490C, T490S, V493T, V493S, V493A, V493R, V493D, V493P, V493G, N597K, N597R, N597A, N597E, N597H, N597G, N597Y, A589V, S599T, N613K, N613R, N613A, N613E, based on amino acid sequence positions of hADAR2-D, and mutations in a homologous ADAR protein corresponding to the above.

[00115] In some embodiments, to reduce editing efficiency, the adenosine deaminase may comprise one or more of the mutations: E488F, E488L, E488W, T490A, T490F, T490Y, T490R, T490K, T490P, T490E, N597F, based on amino acid sequence positions of hADAR2-D, and mutations in a homologous ADAR protein corresponding to the above. In particular embodiments, it can be of interest to use an adenosine deaminase enzyme with reduced efficicay to reduce off-target effects.

[00116] The terms "editing specificity" and "editing preference" are used interchangeably herein to refer to the extent of A-to-I editing at a particular adenosine site in a double-stranded substrate. In some embodiment, the substrate editing preference is determined by the 5' nearest neighbor and/or the 3' nearest neighbor of the target adenosine residue. In some embodiments, the adenosine deaminase has preference for the 5' nearest neighbor of the substrate ranked as U>A>C>G (">" indicates greater preference). In some embodiments, the adenosine deaminase has preference for the 3' nearest neighbor of the substrate ranked as G>C~A>U (">" indicates greater preference; "~" indicates similar preference). In some embodiments, the adenosine deaminase has preference for the 3' nearest neighbor of the substrate ranked as G>C>U~A (">" indicates greater preference; "~" indicates similar preference). In some embodiments, the adenosine deaminase has preference for the 3' nearest neighbor of the substrate ranked as G>C>A>U (">" indicates greater preference). In some embodiments, the adenosine deaminase has preference for the 3' nearest neighbor of the substrate ranked as C~G~A>U (">" indicates greater preference; "~" indicates similar preference). In some embodiments, the adenosine deaminase has preference for a triplet sequence containing the target adenosine residue ranked as

TAG>AAG>CAC>AAT>GAA>GAC (">" indicates greater preference), the center A being the target adenosine residue.

[00117] In some embodiments, the substrate editing preference of an adenosine deaminase is affected by the presence or absence of a nucleic acid binding domain in the adenosine deaminase protein. In some embodiments, to modify substrate editing preference, the deaminase domain is connected with a double-strand RNA binding domain (dsRBD) or a double-strand RNA binding motif (dsRBM). In some embodiments, the dsRBD or dsRBM may be derived from an ADAR protein, such as hADAR1 or hADAR2. In some embodiments, a full length ADAR protein that comprises at least one dsRBD and a deaminase domain is used. In some embodiments, the one or more dsRBM or dsRBD is at the N-terminus of the deaminase domain. In other embodiments, the one or more dsRBM or dsRBD is at the C-terminus of the deaminase domain.

[00118] In some embodiments, the substrate editing preference of an adenosine deaminase is affected by amino acid residues near or in the active center of the enzyme. In some embodiments, to modify substrate editing preference, the adenosine deaminase may comprise one or more of the mutations: G336D, G487R, G487K, G487W, G487Y, E488Q, E488N, T490A, V493A, V493T, V493S, N597K, N597R, A589V, S599T, N613K, N613R, based on amino acid sequence positions of hADAR2-D, and mutations in a homologous ADAR protein corresponding to the above.

[00119] Particularly, in some embodiments, to reduce editing specificity, the adenosine deaminase can comprise one or more of mutations E488Q, V493A, N597K, N613K, based on amino acid sequence positions of hADAR2-D, and mutations in a homologous ADAR protein corresponding to the above. In some embodiments, to increase editing specificity, the adenosine deaminase can comprise mutation T490A.

[00120] In some embodiments, to increase editing preference for target adenosine (A) with an immediate 5' G, such as substrates comprising the triplet sequence GAC, the center A being the target adenosine residue, the adenosine deaminase can comprise one or more of mutations G336D, E488Q, E488N, V493T, V493S, V493A, A589V, N597K, N597R, S599T, N613K, N613R, based on amino acid sequence positions of hADAR2-D, and mutations in a homologous ADAR protein corresponding to the above.

[00121] Particularly, in some embodiments, the adenosine deaminase comprises mutation E488Q or a corresponding mutation in a homologous ADAR protein for editing substrates comprising the following triplet sequences: GAC, GAA, GAU, GAG, CAU, AAU, UAC, the center A being the target adenosine residue.

[00122] In some embodiments, to reduce off-target effects, the adenosine deaminase comprises one or more of mutations at R348, V351, T375, K376, E396, C451, R455, N473, R474, K475, R477, R481, S486, E488, T490, S495, R510, based on amino acid sequence positions of hADAR2-D, and mutations in a homologous ADAR protein corresponding to the above. In some embodiments, the adenosine deaminase comprises mutation at E488 and one or more additional positions selected from R348, V351, T375, K376, E396, C451, R455, N473, R474, K475, R477, R481, S486, T490, S495, R510. In some embodiments, the adenosine deaminase comprises mutation at T375, and optionally at one or more additional positions. In some embodiments, the adenosine deaminase comprises mutation at N473, and optionally at one or more additional positions. In some embodiments, the adenosine deaminase comprises mutation at V351, and optionally at one or more additional positions. In some embodiments, the adenosine deaminase comprises mutation at E488 and T375, and optionally at one or more additional positions. In some embodiments, the adenosine deaminase comprises mutation at E488 and N473, and optionally at one or more additional positions. In some embodiments, the adenosine deaminase comprises mutation E488 and V351, and optionally at one or more additional positions. In some embodiments, the adenosine deaminase comprises mutation at E488 and one or more of T375, N473, and V351.

[00123] In some embodiments, to reduce off-target effects, the adenosine deaminase comprises one or more of mutations selected from R348E, V351L, T375G, T375S, R455G, R455S, R455E, N473D, R474E, K475Q, R477E, R481E, S486T, E488Q, T490A, T490S, S495T, and R510E, based on amino acid sequence positions of hADAR2-D, and mutations in a homologous ADAR protein corresponding to the above. In some embodiments, the adenosine deaminase comprises mutation E488Q and one or more additional mutations selected from R348E, V351L, T375G, T375S, R455G, R455S, R455E, N473D, R474E, K475Q, R477E, R481E, S486T, T490A, T490S, S495T, and R510E. In some embodiments, the adenosine deaminase comprises mutation T375G or T375S, and optionally one or more additional mutations. In some embodiments, the adenosine deaminase comprises mutation N473D, and optionally one or more additional mutations. In some embodiments, the adenosine deaminase comprises mutation V351L, and optionally one or more additional mutations. In some embodiments, the adenosine deaminase comprises mutation E488Q, and T375G or T375G, and optionally one or more additional mutations. In some embodiments, the adenosine deaminase comprises mutation E488Q and N473D, and optionally one or more additional mutations. In some embodiments, the adenosine

deaminase comprises mutation E488Q and V351L, and optionally one or more additional mutations. In some embodiments, the adenosine deaminase comprises mutation E488Q and one or more of T375G/S, N473D and V351L.

[00124] Crystal structures of the human ADAR2 deaminase domain bound to duplex RNA reveal a protein loop that binds the RNA on the 5' side of the modification site. This 5' binding loop is one contributor to substrate specificity differences between ADAR family members. See Wang *et al., Nucleic Acids Res.*, 44(20):9872-9880 (2016), the content of which is incorporated herein by reference in its entirety. In addition, an ADAR2-specific RNA-binding loop was identified near the enzyme active site. See Mathews *et al., Nat. Struct. Mol. Biol.*, 23(5):426-33 (2016), the content of which is incorporated herein by reference in its entirety. In some embodiments, the adenosine deaminase comprises one or more mutations in the RNA binding loop to improve editing specificity and/or efficiency.

[00125] In some embodiments, the adenosine deaminase comprises a mutation at alanine^{454} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the alanine residue at position 454 is replaced by a serine residue (A454S). In some embodiments, the alanine residue at position 454 is replaced by a cysteine residue (A454C). In some embodiments, the alanine residue at position 454 is replaced by an aspartic acid residue (A454D).

[00126] In some embodiments, the adenosine deaminase comprises a mutation at arginine^{455} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the arginine residue at position 455 is replaced by an alanine residue (R455A). In some embodiments, the arginine residue at position 455 is replaced by a valine residue (R455V). In some embodiments, the arginine residue at position 455 is replaced by a histidine residue (R455H). In some embodiments, the arginine residue at position 455 is replaced by a glycine residue (R455G). In some embodiments, the arginine residue at position 455 is replaced by a serine residue (R455S). In some embodiments, the arginine residue at position 455 is replaced by a glutamic acid residue (R455E). In some embodiments, the adenosine deaminase comprises mutation R455C. In some embodiments, the adenosine deaminase comprises mutation R455I. In some embodiments, the adenosine deaminase comprises mutation R455K. In some embodiments, the adenosine deaminase comprises mutation R455L. In some embodiments, the adenosine deaminase comprises mutation R455M. In some embodiments, the adenosine deaminase comprises mutation R455N. In some embodiments, the adenosine deaminase comprises mutation R455Q. In some embodiments, the adenosine deaminase comprises

mutation R455F. In some embodiments, the adenosine deaminase comprises mutation R455W. In some embodiments, the adenosine deaminase comprises mutation R455P. In some embodiments, the adenosine deaminase comprises mutation R455Y. In some embodiments, the adenosine deaminase comprises mutation R455E. In some embodiments, the adenosine deaminase comprises mutation R455D. In some embodiments, the mutations at at R455 described above are further made in combination with a E488Q mutation.

[00127] In some embodiments, the adenosine deaminase comprises a mutation at isoleucine^{456} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the isoleucine residue at position 456 is replaced by a valine residue (I456V). In some embodiments, the isoleucine residue at position 456 is replaced by a leucine residue (I456L). In some embodiments, the isoleucine residue at position 456 is replaced by an aspartic acid residue (I456D).

[00128] In some embodiments, the adenosine deaminase comprises a mutation at phenylalanine^{457} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the phenylalanine residue at position 457 is replaced by a tyrosine residue (F457Y). In some embodiments, the phenylalanine residue at position 457 is replaced by an arginine residue (F457R). In some embodiments, the phenylalanine residue at position 457 is replaced by a glutamic acid residue (F457E).

[00129] In some embodiments, the adenosine deaminase comprises a mutation at serine^{458} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the serine residue at position 458 is replaced by a valine residue (S458V). In some embodiments, the serine residue at position 458 is replaced by a phenylalanine residue (S458F). In some embodiments, the serine residue at position 458 is replaced by a proline residue (S458P). In some embodiments, the adenosine deaminase comprises mutation S458I. In some embodiments, the adenosine deaminase comprises mutation S458L. In some embodiments, the adenosine deaminase comprises mutation S458M. In some embodiments, the adenosine deaminase comprises mutation S458C. In some embodiments, the adenosine deaminase comprises mutation S458A. In some embodiments, the adenosine deaminase comprises mutation S458G. In some embodiments, the adenosine deaminase comprises mutation S458T. In some embodiments, the adenosine deaminase comprises mutation S458Y. In some embodiments, the adenosine deaminase comprises mutation S458W. In some embodiments, the adenosine deaminase comprises mutation S458Q. In some embodiments, the adenosine deaminase comprises mutation S458N. In some embodiments, the adenosine deaminase comprises

mutation S458H. In some embodiments, the adenosine deaminase comprises mutation S458E. In some embodiments, the adenosine deaminase comprises mutation S458D. In some embodiments, the adenosine deaminase comprises mutation S458K. In some embodiments, the adenosine deaminase comprises mutation S458R. In some embodiments, the mutations at S458 described above are further made in combination with a E488Q mutation.

[00130] In some embodiments, the adenosine deaminase comprises a mutation at proline^{459} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the proline residue at position 459 is replaced by a cysteine residue (P459C). In some embodiments, the proline residue at position 459 is replaced by a histidine residue (P459H). In some embodiments, the proline residue at position 459 is replaced by a tryptophan residue (P459W).

[00131] In some embodiments, the adenosine deaminase comprises a mutation at histidine^{460} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the histidine residue at position 460 is replaced by an arginine residue (H460R). In some embodiments, the histidine residue at position 460 is replaced by an isoleucine residue (H460I). In some embodiments, the histidine residue at position 460 is replaced by a proline residue (H460P). In some embodiments, the adenosine deaminase comprises mutation H460L. In some embodiments, the adenosine deaminase comprises mutation H460V. In some embodiments, the adenosine deaminase comprises mutation H460F. In some embodiments, the adenosine deaminase comprises mutation H460M. In some embodiments, the adenosine deaminase comprises mutation H460C. In some embodiments, the adenosine deaminase comprises mutation H460A. In some embodiments, the adenosine deaminase comprises mutation H460G. In some embodiments, the adenosine deaminase comprises mutation H460T. In some embodiments, the adenosine deaminase comprises mutation H460S. In some embodiments, the adenosine deaminase comprises mutation H460Y. In some embodiments, the adenosine deaminase comprises mutation H460W. In some embodiments, the adenosine deaminase comprises mutation H460Q. In some embodiments, the adenosine deaminase comprises mutation H460N. In some embodiments, the adenosine deaminase comprises mutation H460E. In some embodiments, the adenosine deaminase comprises mutation H460D. In some embodiments, the adenosine deaminase comprises mutation H460K. In some embodiments, the mutations at H460 described above are further made in combination with a E488Q mutation.

[00132] In some embodiments, the adenosine deaminase comprises a mutation at proline^{462} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the proline residue at position 462 is replaced by a serine residue (P462S). In some embodiments, the proline residue at position 462 is replaced by a tryptophan residue (P462W). In some embodiments, the proline residue at position 462 is replaced by a glutamic acid residue (P462E).

[00133] In some embodiments, the adenosine deaminase comprises a mutation at aspartic acid^{469} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the aspartic acid residue at position 469 is replaced by a glutamine residue (D469Q). In some embodiments, the aspartic acid residue at position 469 is replaced by a serine residue (D469S). In some embodiments, the aspartic acid residue at position 469 is replaced by a tyrosine residue (D469Y).

[00134] In some embodiments, the adenosine deaminase comprises a mutation at arginine^{470} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the arginine residue at position 470 is replaced by an alanine residue (R470A). In some embodiments, the arginine residue at position 470 is replaced by an isoleucine residue (R470I). In some embodiments, the arginine residue at position 470 is replaced by an aspartic acid residue (R470D).

[00135] In some embodiments, the adenosine deaminase comprises a mutation at histidine^{471} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the histidine residue at position 471 is replaced by a lysine residue (H471K). In some embodiments, the histidine residue at position 471 is replaced by a threonine residue (H471T). In some embodiments, the histidine residue at position 471 is replaced by a valine residue (H471V).

[00136] In some embodiments, the adenosine deaminase comprises a mutation at proline^{472} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the proline residue at position 472 is replaced by a lysine residue (P472K). In some embodiments, the proline residue at position 472 is replaced by a threonine residue (P472T). In some embodiments, the proline residue at position 472 is replaced by an aspartic acid residue (P472D).

[00137] In some embodiments, the adenosine deaminase comprises a mutation at asparagine^{473} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the asparagine residue at position 473 is replaced by an arginine residue (N473R). In some embodiments, the asparagine residue at

position 473 is replaced by a tryptophan residue (N473W). In some embodiments, the asparagine residue at position 473 is replaced by a proline residue (N473P). In some embodiments, the asparagine residue at position 473 is replaced by an aspartic acid residue (N473D).

[00138] In some embodiments, the adenosine deaminase comprises a mutation at arginine^{474} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the arginine residue at position 474 is replaced by a lysine residue (R474K). In some embodiments, the arginine residue at position 474 is replaced by a glycine residue (R474G). In some embodiments, the arginine residue at position 474 is replaced by an aspartic acid residue (R474D). In some embodiments, the arginine residue at position 474 is replaced by a glutamic acid residue (R474E).

[00139] In some embodiments, the adenosine deaminase comprises a mutation at lysine^{475} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the lysine residue at position 475 is replaced by a glutamine residue (K475Q). In some embodiments, the lysine residue at position 475 is replaced by an asparagine residue (K475N). In some embodiments, the lysine residue at position 475 is replaced by an aspartic acid residue (K475D).

[00140] In some embodiments, the adenosine deaminase comprises a mutation at alanine^{476} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the alanine residue at position 476 is replaced by a serine residue (A476S). In some embodiments, the alanine residue at position 476 is replaced by an arginine residue (A476R). In some embodiments, the alanine residue at position 476 is replaced by a glutamic acid residue (A476E).

[00141] In some embodiments, the adenosine deaminase comprises a mutation at arginine^{477} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the arginine residue at position 477 is replaced by a lysine residue (R477K). In some embodiments, the arginine residue at position 477 is replaced by a threonine residue (R477T). In some embodiments, the arginine residue at position 477 is replaced by a phenylalanine residue (R477F). In some embodiments, the arginine residue at position 474 is replaced by a glutamic acid residue (R477E).

[00142] In some embodiments, the adenosine deaminase comprises a mutation at glycine^{478} of the hADAR2-D amino acid sequence, or a corresponding position in a

homologous ADAR protein. In some embodiments, the glycine residue at position 478 is replaced by an alanine residue (G478A). In some embodiments, the glycine residue at position 478 is replaced by an arginine residue (G478R). In some embodiments, the glycine residue at position 478 is replaced by a tyrosine residue (G478Y). In some embodiments, the adenosine deaminase comprises mutation G478I. In some embodiments, the adenosine deaminase comprises mutation G478L. In some embodiments, the adenosine deaminase comprises mutation G478V. In some embodiments, the adenosine deaminase comprises mutation G478F. In some embodiments, the adenosine deaminase comprises mutation G478M. In some embodiments, the adenosine deaminase comprises mutation G478C. In some embodiments, the adenosine deaminase comprises mutation G478P. In some embodiments, the adenosine deaminase comprises mutation G478T. In some embodiments, the adenosine deaminase comprises mutation G478S. In some embodiments, the adenosine deaminase comprises mutation G478W. In some embodiments, the adenosine deaminase comprises mutation G478Q. In some embodiments, the adenosine deaminase comprises mutation G478N. In some embodiments, the adenosine deaminase comprises mutation G478H. In some embodiments, the adenosine deaminase comprises mutation G478E. In some embodiments, the adenosine deaminase comprises mutation G478D. In some embodiments, the adenosine deaminase comprises mutation G478K. In some embodiments, the mutations at G478 described above are further made in combination with a E488Q mutation.

[00143] In some embodiments, the adenosine deaminase comprises a mutation at glutamine^{479} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the glutamine residue at position 479 is replaced by an asparagine residue (Q479N). In some embodiments, the glutamine residue at position 479 is replaced by a serine residue (Q479S). In some embodiments, the glutamine residue at position 479 is replaced by a proline residue (Q479P).

[00144] In some embodiments, the adenosine deaminase comprises a mutation at arginine^{348} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the arginine residue at position 348 is replaced by an alanine residue (R348A). In some embodiments, the arginine residue at position 348 is replaced by a glutamic acid residue (R348E).

[00145] In some embodiments, the adenosine deaminase comprises a mutation at valine^{351} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the valine residue at position 351 is

replaced by a leucine residue (V351L). In some embodiments, the adenosine deaminase comprises mutation V351Y. In some embodiments, the adenosine deaminase comprises mutation V351M. In some embodiments, the adenosine deaminase comprises mutation V351T. In some embodiments, the adenosine deaminase comprises mutation V351G. In some embodiments, the adenosine deaminase comprises mutation V351A. In some embodiments, the adenosine deaminase comprises mutation V351F. In some embodiments, the adenosine deaminase comprises mutation V351E. In some embodiments, the adenosine deaminase comprises mutation V351I. In some embodiments, the adenosine deaminase comprises mutation V351C. In some embodiments, the adenosine deaminase comprises mutation V351H. In some embodiments, the adenosine deaminase comprises mutation V351P. In some embodiments, the adenosine deaminase comprises mutation V351S. In some embodiments, the adenosine deaminase comprises mutation V351K. In some embodiments, the adenosine deaminase comprises mutation V351N. In some embodiments, the adenosine deaminase comprises mutation V351W. In some embodiments, the adenosine deaminase comprises mutation V351Q. In some embodiments, the adenosine deaminase comprises mutation V351D. In some embodiments, the adenosine deaminase comprises mutation V351R. In some embodiments, the mutations at V351 described above are further made in combination with a E488Q mutation.

[00146] In some embodiments, the adenosine deaminase comprises a mutation at threonine^{375} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the threonine residue at position 375 is replaced by a glycine residue (T375G). In some embodiments, the threonine residue at position 375 is replaced by a serine residue (T375S). In some embodiments, the adenosine deaminase comprises mutation T375H. In some embodiments, the adenosine deaminase comprises mutation T375Q. In some embodiments, the adenosine deaminase comprises mutation T375C. In some embodiments, the adenosine deaminase comprises mutation T375N. In some embodiments, the adenosine deaminase comprises mutation T375M. In some embodiments, the adenosine deaminase comprises mutation T375A. In some embodiments, the adenosine deaminase comprises mutation T375W. In some embodiments, the adenosine deaminase comprises mutation T375V. In some embodiments, the adenosine deaminase comprises mutation T375R. In some embodiments, the adenosine deaminase comprises mutation T375E. In some embodiments, the adenosine deaminase comprises mutation T375K. In some embodiments, the adenosine deaminase comprises mutation T375F. In some embodiments, the adenosine deaminase comprises mutation

T375I. In some embodiments, the adenosine deaminase comprises mutation T375D. In some embodiments, the adenosine deaminase comprises mutation T375P. In some embodiments, the adenosine deaminase comprises mutation T375L. In some embodiments, the adenosine deaminase comprises mutation T375Y. In some embodiments, the mutations at T375Y described above are further made in combination with an E488Q mutation.

[00147] In some embodiments, the adenosine deaminase comprises a mutation at arginine^{481} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the arginine residue at position 481 is replaced by a glutamic acid residue (R481E).

[00148] In some embodiments, the adenosine deaminase comprises a mutation at serine^{486} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the serine residue at position 486 is replaced by a threonine residue (S486T).

[00149] In some embodiments, the adenosine deaminase comprises a mutation at threonine^{490} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the threonine residue at position 490 is replaced by an alanine residue (T490A). In some embodiments, the threonine residue at position 490 is replaced by a serine residue (T490S).

[00150] In some embodiments, the adenosine deaminase comprises a mutation at serine^{495} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the serine residue at position 495 is replaced by a threonine residue (S495T).

[00151] In some embodiments, the adenosine deaminase comprises a mutation at arginine^{510} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the arginine residue at position 510 is replaced by a glutamine residue (R510Q). In some embodiments, the arginine residue at position 510 is replaced by an alanine residue (R510A). In some embodiments, the arginine residue at position 510 is replaced by a glutamic acid residue (R510E).

[00152] In some embodiments, the adenosine deaminase comprises a mutation at glycine^{593} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the glycine residue at position 593 is replaced by an alanine residue (G593A). In some embodiments, the glycine residue at position 593 is replaced by a glutamic acid residue (G593E).

[00153] In some embodiments, the adenosine deaminase comprises a mutation at lysine^{594} of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the lysine residue at position 594 is replaced by an alanine residue (K594A).

[00154] In some embodiments, the adenosine deaminase comprises a mutation at any one or more of positions A454, R455, I456, F457, S458, P459, H460, P462, D469, R470, H471, P472, N473, R474, K475, A476, R477, G478, Q479, R348, R510, G593, K594 of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein.

[00155] In some embodiments, the adenosine deaminase comprises any one or more of mutations A454S, A454C, A454D, R455A, R455V, R455H, I456V, I456L, I456D, F457Y, F457R, F457E, S458V, S458F, S458P, P459C, P459H, P459W, H460R, H460I, H460P, P462S, P462W, P462E, D469Q, D469S, D469Y, R470A, R470I, R470D, H471K, H471T, H471V, P472K, P472T, P472D, N473R, N473W, N473P, R474K, R474G, R474D, K475Q, K475N, K475D, A476S, A476R, A476E, R477K, R477T, R477F, G478A, G478R, G478Y, Q479N, Q479S, Q479P, R348A, R510Q, R510A, G593A, G593E, K594A of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein.

[00156] In some embodiments, the adenosine deaminase comprises a mutation at any one or more of positions T375, V351, G478, S458, H460 of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein, optionally in combination a mutation at E488. In some embodiments, the adenosine deaminase comprises one or more of mutations selected from T375G, T375C, T375H, T375Q, V351M, V351T, V351Y, G478R, S458F, H460I, optionally in combination with E488Q.

[00157] In some embodiments, the adenosine deaminase comprises one or more of mutations selected from T375H, T375Q, V351M, V351Y, H460P, optionally in combination with E488Q.

[00158] In some embodiments, the adenosine deaminase comprises mutations T375S and S458F, optionally in combination with E488Q.

[00159] In some embodiments, the adenosine deaminase comprises a mutation at two or more of positions T375, N473, R474, G478, S458, P459, V351, R455, R455, T490, R348, Q479 of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein, optionally in combination a mutation at E488. In some embodiments, the adenosine deaminase comprises two or more of mutations selected from T375G, T375S,

N473D, R474E, G478R, S458F, P459W, V351L, R455G, R455S, T490A, R348E, Q479P, optionally in combination with E488Q.

[00160] In some embodiments, the adenosine deaminase comprises mutations T375G and V351L. In some embodiments, the adenosine deaminase comprises mutations T375G and R455G. In some embodiments, the adenosine deaminase comprises mutations T375G and R455S. In some embodiments, the adenosine deaminase comprises mutations T375G and T490A. In some embodiments, the adenosine deaminase comprises mutations T375G and R348E. In some embodiments, the adenosine deaminase comprises mutations T375S and V351L. In some embodiments, the adenosine deaminase comprises mutations T375S and R455G. In some embodiments, the adenosine deaminase comprises mutations T375S and R455S. In some embodiments, the adenosine deaminase comprises mutations T375S and T490A. In some embodiments, the adenosine deaminase comprises mutations T375S and R348E. In some embodiments, the adenosine deaminase comprises mutations N473D and V351L. In some embodiments, the adenosine deaminase comprises mutations N473D and R455G. In some embodiments, the adenosine deaminase comprises mutations N473D and R455S. In some embodiments, the adenosine deaminase comprises mutations N473D and T490A. In some embodiments, the adenosine deaminase comprises mutations N473D and R348E. In some embodiments, the adenosine deaminase comprises mutations R474E and V351L. In some embodiments, the adenosine deaminase comprises mutations R474E and R455G. In some embodiments, the adenosine deaminase comprises mutations R474E and R455S. In some embodiments, the adenosine deaminase comprises mutations R474E and T490A. In some embodiments, the adenosine deaminase comprises mutations R474E and R348E. In some embodiments, the adenosine deaminase comprises mutations S458F and T375G. In some embodiments, the adenosine deaminase comprises mutations S458F and T375S. In some embodiments, the adenosine deaminase comprises mutations S458F and N473D. In some embodiments, the adenosine deaminase comprises mutations S458F and R474E. In some embodiments, the adenosine deaminase comprises mutations S458F and G478R. In some embodiments, the adenosine deaminase comprises mutations G478R and T375G. In some embodiments, the adenosine deaminase comprises mutations G478R and T375S. In some embodiments, the adenosine deaminase comprises mutations G478R and N473D. In some embodiments, the adenosine deaminase comprises mutations G478R and R474E. In some embodiments, the adenosine deaminase comprises mutations P459W and T375G. In some embodiments, the adenosine deaminase comprises mutations P459W and T375S. In some embodiments, the adenosine deaminase comprises mutations P459W

and N473D. In some embodiments, the adenosine deaminase comprises mutations P459W and R474E. In some embodiments, the adenosine deaminase comprises mutations P459W and G478R. In some embodiments, the adenosine deaminase comprises mutations P459W and S458F. In some embodiments, the adenosine deaminase comprises mutations Q479P and T375G. In some embodiments, the adenosine deaminase comprises mutations Q479P and T375S. In some embodiments, the adenosine deaminase comprises mutations Q479P and N473D. In some embodiments, the adenosine deaminase comprises mutations Q479P and R474E. In some embodiments, the adenosine deaminase comprises mutations Q479P and G478R. In some embodiments, the adenosine deaminase comprises mutations Q479P and S458F. In some embodiments, the adenosine deaminase comprises mutations Q479P and P459W. All mutations described in this paragraph may also further be made in cominbation with a E488Q mutations.

[00161] In some embodiments, the adenosine deaminase comprises a mutation at any one or more of positions K475, Q479, P459, G478, S458of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein, optionally in combination a mutation at E488. In some embodiments, the adenosine deaminase comprises one or more of mutations selected from K475N, Q479N, P459W, G478R, S458P, S458F, optionally in combination with E488Q.

[00162] In some embodiments, the adenosine deaminase comprises a mutation at any one or more of positions T375, V351, R455, H460, A476 of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein, optionally in combination a mutation at E488. In some embodiments, the adenosine deaminase comprises one or more of mutations selected from T375G, T375C, T375H, T375Q, V351M, V351T, V351Y, R455H, H460P, H460I, A476E, optionally in combination with E488Q.

[00163] In certain embodiments, improvement of editing and reduction of off-target modification is achieved by chemical modification of gRNAs. gRNAs which are chemically modified as exemplified in Vogel et al. (2014), Angew Chem Int Ed, 53:6267-6271, doi:10.1002/anie.201402634 (incorporated herein by reference in its entirety) reduce off-target activity and improve on-target efficiency. 2′-O-methyl and phosphothioate modified guide RNAs in general improve editing efficiency in cells.

[00164] ADAR has been known to demonstrate a preference for neighboring nucleotides on either side of the edited A (www.nature.com/nsmb/journal/v23/n5/full/nsmb.3203.html, Matthews et al. (2017), Nature Structural Mol Biol, 23(5): 426-433, incorporated herein by

reference in its entirety). Accordingly, in certain embodiments, the gRNA, target, and/or ADAR is selected optimized for motif preference.

[00165] Intentional mismatches have been demonstrated in vitro to allow for editing of non-preferred motifs (https://academic.oup.com/nar/article-lookup/doi/10.1093/nar/gku272; Schneider et al (2014), Nucleic Acid Res, 42(10):e87); Fukuda et al. (2017), Scienticic Reports, 7, doi:10.1038/srep41478, incorporated herein by reference in its entirety). Accordingly, in certain embodiments, to enhance RNA editing efficiency on non-preferred 5' or 3' neighboring bases, intentional mismatches in neighboring bases are introduced.

[00166] As opposite Cs in the targeting window of the ADAR deaminase domain may be preferentially edited over other bases, while As base-paired with Us within a few bases of the targeted base may have low levels of editing. Accordingly, multiple As in the activity window of Cpf1-ADAR systemt could be specified for editing by mismatching all As to be edited with Cs. Accordingly, in certain embodiments, multiple A:C mismatches in the activity window are designed to create multiple A:I edits. In certain embodiments, to suppress potential off-target editing in the activity window, non-target As are paired with As or Gs.

[00167] In some embodiments, the adenosine deaminase comprises the wild-type amino acid sequence of hADAR1-D (e.g., MGSGGGGSEGAPKKKRKVGSSLGTGNRCVKGDSLSLKGE TVNDCHAEIISRRGFIRFLYSELMKYNSQTAKDSIFEPAKGGEKLQIKKTVSFHLYIS TAPCGDGALFDKSCSDRAMESTESRHYPVFENPKQGKLRTKVENGQGTIPVESSDI VPTWDGIRLGERLRTMSCSDKILRWNVLGLQGALLTHFLQPIYLKSVTLGYLFSQG HLTRAICCRVTRDGSAFEDGLRHPFIVNHPKVGRVSIYDSKRQSGKTKETSVNWCL ADGYDLEILDGTRGTVDGPRNELSRVSKKNIFLLFKKLCSFRYRRDLLRLSYGEAK KAARDYETAKNYFKKGLKDMGYGNWISKPQEEKNF* (SEQ ID NO:36)). In some embodiments, the adenosine deaminase comprises one or more mutations in the hADAR1-D sequence, such that the editing efficiency, and/or substrate editing preference of hADAR1-D is changed according to specific needs.

[00168] In some embodiments, the adenosine deaminase comprises a mutation at Glycine^{1007} of the hADAR1-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the glycine residue at position 1007 is replaced by a non-polar amino acid residue with relatively small side chains. For example, in some embodiments, the glycine residue at position 1007 is replaced by an alanine residue (G1007A). In some embodiments, the glycine residue at position 1007 is replaced by a

valine residue (G1007V). In some embodiments, the glycine residue at position 1007 is replaced by an amino acid residue with relatively large side chains. In some embodiments, the glycine residue at position 1007 is replaced by an arginine residue (G1007R). In some embodiments, the glycine residue at position 1007 is replaced by a lysine residue (G1007K). In some embodiments, the glycine residue at position 1007 is replaced by a tryptophan residue (G1007W). In some embodiments, the glycine residue at position 1007 is replaced by a tyrosine residue (G1007Y). Additionally, in other embodiments, the glycine residue at position 1007 is replaced by a leucine residue (G1007L). In other embodiments, the glycine residue at position 1007 is replaced by a threonine residue (G1007T). In other embodiments, the glycine residue at position 1007 is replaced by a serine residue (G1007S).

[00169] In some embodiments, the adenosine deaminase comprises a mutation at glutamic acid^{1008} of the hADAR1-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the glutamic acid residue at position 1008 is replaced by a polar amino acid residue having a relatively large side chain. In some embodiments, the glutamic acid residue at position 1008 is replaced by a glutamine residue (E1008Q). In some embodiments, the glutamic acid residue at position 1008 is replaced by a histidine residue (E1008H). In some embodiments, the glutamic acid residue at position 1008 is replaced by an arginine residue (E1008R). In some embodiments, the glutamic acid residue at position 1008 is replaced by a lysine residue (E1008K). In some embodiments, the glutamic acid residue at position 1008 is replaced by a nonpolar or small polar amino acid residue. In some embodiments, the glutamic acid residue at position 1008 is replaced by a phenylalanine residue (E1008F). In some embodiments, the glutamic acid residue at position 1008 is replaced by a tryptophan residue (E1008W). In some embodiments, the glutamic acid residue at position 1008 is replaced by a glycine residue (E1008G). In some embodiments, the glutamic acid residue at position 1008 is replaced by an isoleucine residue (E1008I). In some embodiments, the glutamic acid residue at position 1008 is replaced by a valine residue (E1008V). In some embodiments, the glutamic acid residue at position 1008 is replaced by a proline residue (E1008P). In some embodiments, the glutamic acid residue at position 1008 is replaced by a serine residue (E1008S). In other embodiments, the glutamic acid residue at position 1008 is replaced by an asparagine residue (E1008N). In other embodiments, the glutamic acid residue at position 1008 is replaced by an alanine residue (E1008A). In other embodiments, the glutamic acid residue at position 1008 is replaced by a Methionine residue (E1008M). In some embodiments, the glutamic acid residue at position 1008 is replaced by a leucine residue (E1008L).

[00170] In some embodiments, to improve editing efficiency, the adenosine deaminase may comprise one or more of the mutations: E1007S, E1007A, E1007V, E1008Q, E1008R, E1008H, E1008M, E1008N, E1008K, based on amino acid sequence positions of hADAR1-D, and mutations in a homologous ADAR protein corresponding to the above.

[00171] In some embodiments, to reduce editing efficiency, the adenosine deaminase may comprise one or more of the mutations: E1007R, E1007K, E1007Y, E1007L, E1007T, E1008G, E1008I, E1008P, E1008V, E1008F, E1008W, E1008S, E1008N, E1008K, based on amino acid sequence positions of hADAR1-D, and mutations in a homologous ADAR protein corresponding to the above.

[00172] In some embodiments, the substrate editing preference, efficiency and/or selectivity of an adenosine deaminase is affected by amino acid residues near or in the active center of the enzyme. In some embodiments, the adenosine deaminase comprises a mutation at the glutamic acid 1008 position in hADAR1-D sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the mutation is E1008R, or a corresponding mutation in a homologous ADAR protein. In some embodiments, the E1008R mutant has an increased editing efficiency for target adenosine residue that has a mismatched G residue on the opposite strand.

[00173] In some embodiments, the adenosine deaminase protein further comprises or is connected to one or more double-stranded RNA (dsRNA) binding motifs (dsRBMs) or domains (dsRBDs) for recognizing and binding to double-stranded nucleic acid substrates. In some embodiments, the interaction between the adenosine deaminase and the double–stranded substrate is mediated by one or more additional protein factor(s), including a CRISPR/CAS protein factor. In some embodiments, the interaction between the adenosine deaminase and the double-stranded substrate is further mediated by one or more nucleic acid component(s), including a guide RNA.

[00174] According to the present invention, the substrate of the adenosine deaminase is an RNA/DNA heteroduplex formed upon binding of the guide molecule to its DNA target which then forms the CRISPR-Cas complex with the CRISPR-Cas enzyme. The RNA/DNA or DNA/RNA heteroduplex is also referred to herein as the "RNA/DNA hybrid", "DNA/RNA hybrid" or "double-stranded substrate". The particular features of the guide molecule and CRISPR-Cas enzyme are detailed below.

[00175] The term "editing selectivity" as used herein refers to the fraction of all sites on a double-stranded substrate that is edited by an adenosine deaminase. Without being bound

by theory, it is contemplated that editing selectivity of an adenosine deaminase is affected by the double-stranded substrate's length and secondary structures, such as the presence of mismatched bases, bulges and/or internal loops.

[00176] In some embodiments, when the substrate is a perfectly base-paired duplex longer than 50 bp, the adenosine deaminase may be able to deaminate multiple adenosine residues within the duplex (e.g., 50% of all adenosine residues). In some embodiments, when the substrate is shorter than 50 bp, the editing selectivity of an adenosine deaminase is affected by the presence of a mismatch at the target adenosine site. Particularly, in some embodiments, adenosine (A) residue having a mismatched cytidine (C) residue on the opposite strand is deaminated with high efficiency. In some embodiments, adenosine (A) residue having a mismatched guanosine (G) residue on the opposite strand is skipped without editing.

Modified Adenosine Deaminase Having C-to U Deamination Activity

[00177] In certain example embodiments, directed evolution may be used to design modified ADAR proteins capable of catalyzing additional reactions besides deamination of an adenine to a hypoxanthine. For example, the modified ADAR protein may be capable of catalyzing deamination of a cytidine to a uracil. While not bound by a particular theory, mutations that improve C to U activity may alter the shape of the binding pocket to be more amenable to the smaller cytidine base.

[00178] In some embodiments, the modified adenosine deaminase having C-to-U deamination activity comprises a mutation at any one or more of positions V351, T375, R455, and E488 of the hADAR2-D amino acid sequence, or a corresponding position in a homologous ADAR protein. In some embodiments, the adenosine deaminase comprises mutation E488Q. In some embodiments, the adenosine deaminase comprises one or more of mutations selected from V351I, V351L, V351F, V351M, V351C, V351A, V351G, V351P, V351T, V351S, V351Y, V351W, V351Q, V351N, V351H, V351E, V351D, V351K, V351R, T375I, T375L, T375V, T375F, T375M, T375C, T375A, T375G, T375P, T375S, T375Y, T375W, T375Q, T375N, T375H, T375E, T375D, T375K, T375R, R455I, R455L, R455V, R455F, R455M, R455C, R455A, R455G, R455P, R455T, R455S, R455Y, R455W, R455Q, R455N, R455H, R455E, R455D, R455K. In some embodiments, the adenosine deaminase comprises mutation E488Q, and further comprises one or more of mutations selected from V351I, V351L, V351F, V351M, V351C, V351A, V351G, V351P, V351T, V351S, V351Y, V351W, V351Q, V351N, V351H, V351E, V351D, V351K, V351R, T375I, T375L, T375V, T375F, T375M, T375C, T375A, T375G, T375P, T375S, T375Y,

T375W, T375Q, T375N, T375H, T375E, T375D, T375K, T375R, R455I, R455L, R455V, R455F, R455M, R455C, R455A, R455G, R455P, R455T, R455S, R455Y, R455W, R455Q, R455N, R455H, R455E, R455D, R455K.

[00179] In connection with the aforementioned modified ADAR protein having C-to-U deamination activity, the invention described herein also relates to a method for deaminating a C in a target locus of interest, comprising delivering to said target locus of interest: (a) a Cpf1 nickase protein or a catalytically inactive Cpf1 protein; (b) a guide molecule which comprises a guide sequence linked to a direct repeat sequence; and (c) a modified ADAR protein having C-to-U deamination activity or catalytic domain thereof;

wherein said modified ADAR protein or catalytic domain thereof is covalently or non-covalently linked to said Cpf1 protein or said guide molecule or is adapted to link thereto after delivery;

wherein guide molecule forms a complex with said Cpf1 protein and directs said complex to bind a first DNA strand at said target locus of interest;

wherein said guide sequence is capable of hybridizing with a target sequence comprising said C within said first DNA strand to form a heteroduplex;

wherein, optionally, said guide sequence comprises a non-pairing A or U at a position corresponding to said C resulting in a mismatch in the heteroduplex formed;

wherein, optionally, said Cpf1 protein is a Cpf1 nickase which nicks a second DNA strand at said target locus of interest displaced by formation of said heteroduplex; and

wherein said modified ADAR protein or catalytic domain thereof deaminates said C in said RNA heteroduplex.

[00180] In connection with the aforementioned modified ADAR protein having C-to-U deamination activity, the invention described herein further relates to an engineered, non-naturally occurring system suitable for deaminating a C in a target locus of interest, comprising: (a) a guide molecule which comprises a guide sequence linked to a direct repeat sequence, or a nucleotide sequence encoding said guide molecule; (b) a Cpf1 nickase protein or a catalytically inactive Cpf1 protein, or a nucleotide sequence encoding said Cpf1 protein; (c) a modified ADAR protein having C-to-U deamination activity or catalytic domain thereof, or a nucleotide sequence encoding said modified ADAR protein or catalytic domain thereof;

wherein said modified ADAR protein or catalytic domain thereof is covalently or non-covalently linked to said Cpf1 protein or said guide molecule or is adapted to link thereto after delivery;

wherein said guide sequence is capable of hybridizing with a target sequence comprising a C on a first DNA strand at said target locus to form a heteroduplex;

wherein, optionally, said guide sequence comprises a non-pairing A or U at a position corresponding to said C resulting in a mismatch in the heteroduplex formed;

wherein, optionally, said Cpf1 protein is a Cpf1 nickase capable of nicking a second DNA strand complementary to said first DNA strand;

wherein, optionally, the system is a vector system comprising one or more vectors comprising: (a) a first regulatory element operably linked to a nucleotide sequence encoding said guide molecule which comprises said guide sequence, (b) a second regulatory element operably linked to a nucleotide sequence encoding said Cpf1 protein; and (c) a nucleotide sequence encoding a modified ADAR protein having C-to-U deamination activity or catalytic domain thereof which is under control of said first or second regulatory element or operably linked to a third regulatory element;

wherein, if said nucleotide sequence encoding a modified ADAR protein or catalytic domain thereof is operably linked to a third regulatory element, said modified ADAR protein or catalytic domain thereof is adapted to link to said guide molecule or said Cpf1 protein after expression;

wherein components (a), (b) and (c) are located on the same or different vectors of the system, optionally wherein said first, second, and/or third regulatory element is an inducible promoter.

[00181] CRISPR-Cas Protein and Guide

[00182] In the methods and systems of the present invention use is made of a CRISPR-Cas protein and corresponding guide molecule. More particularly, the CRISPR-Cas protein is a class 2 CRISPR-Cas protein. In certain embodiments, said CRISPR-Cas protein Cpf1. The CRISPR-Cas system does not require the generation of customized proteins to target specific sequences but rather a single Cas protein can be programmed by guide molecule to recognize a specific nucleic acid target, in other words the Cas enzyme protein can be recruited to a specific nucleic acid target locus of interest using said guide molecule.

[00183] *Guide molecule*

[00184] The guide molecule or guide RNA of a Class 2 type V CRISPR-Cas protein comprises a tracr-mate sequence (encompassing a "direct repeat" in the context of an endogenous CRISPR system) and a guide sequence (also referred to as a "spacer" in the context of an endogenous CRISPR system). Indeed, in contrast to the type II CRISPR-Cas proteins, the CRISPR-Cas Cpf1 protein does not rely on the presence of a tracr sequence. In

some embodiments, the CRISPR-Cas system or complex as described herein does not comprise and/or does not rely on the presence of a tracr sequence (e.g. if the Cas protein is Cpf1). In certain embodiments, the guide molecule may comprise, consist essentially of, or consist of a direct repeat sequence fused or linked to a guide sequence or spacer sequence.

[00185] In general, a CRISPR system is characterized by elements that promote the formation of a CRISPR complex at the site of a target sequence. In the context of formation of a CRISPR complex, "target sequence" refers to a sequence to which a guide sequence is designed to have complementarity, where hybridization between a target DNA sequence and a guide sequence promotes the formation of a CRISPR complex.

[00186] The terms "guide molecule" and "guide RNA" are used interchangeably herein to refer to RNA-based molecules that are capable of forming a complex with a CRISPR-Cas protein and comprises a guide sequence having sufficient complementarity with a target nucleic acid sequence to hybridize with the target nucleic acid sequence and direct sequence-specific binding of the complex to the target nucleic acid sequence. The guide molecule or guide RNA specifically encompasses RNA-based molecules having one or more chemically modifications (e.g., by chemical linking two ribonucleotides or by replacement of one or more ribonucleotides with one or more deoxyribonucleotides), as described herein.

[00187] As used herein, the term "guide sequence" in the context of a CRISPR-Cas system, comprises any polynucleotide sequence having sufficient complementarity with a target nucleic acid sequence to hybridize with the target nucleic acid sequence and direct sequence-specific binding of a nucleic acid-targeting complex to the target nucleic acid sequence. In the context of the present invention the target nucleic acid sequence or target sequence is the sequence comprising the target adenosine to be deaminated also referred to herein as the "target adenosine". In some embodiments, except for the intended dA-C mismatch, the degree of complementarity, when optimally aligned using a suitable alignment algorithm, is about or more than about 50%, 60%, 75%, 80%, 85%, 90%, 95%, 97.5%, 99%, or more. Optimal alignment may be determined with the use of any suitable algorithm for aligning sequences, non-limiting example of which include the Smith-Waterman algorithm, the Needleman-Wunsch algorithm, algorithms based on the Burrows-Wheeler Transform (e.g., the Burrows Wheeler Aligner), ClustalW, Clustal X, BLAT, Novoalign (Novocraft Technologies; available at www.novocraft.com), ELAND (Illumina, San Diego, CA), SOAP (available at soap.genomics.org.cn), and Maq (available at maq.sourceforge.net). The ability of a guide sequence (within a nucleic acid-targeting guide

RNA) to direct sequence-specific binding of a nucleic acid -targeting complex to a target nucleic acid sequence may be assessed by any suitable assay. For example, the components of a nucleic acid-targeting CRISPR system sufficient to form a nucleic acid-targeting complex, including the guide sequence to be tested, may be provided to a host cell having the corresponding target nucleic acid sequence, such as by transfection with vectors encoding the components of the nucleic acid-targeting complex, followed by an assessment of preferential targeting (e.g., cleavage) within the target nucleic acid sequence, such as by Surveyor assay as described herein. Similarly, cleavage of a target nucleic acid sequence (or a sequence in the vicinity thereof) may be evaluated in a test tube by providing the target nucleic acid sequence, components of a nucleic acid-targeting complex, including the guide sequence to be tested and a control guide sequence different from the test guide sequence, and comparing binding or rate of cleavage at or in the vicinity of the target sequence between the test and control guide sequence reactions. Other assays are possible, and will occur to those skilled in the art. A guide sequence, and hence a nucleic acid-targeting guide RNA may be selected to target any target nucleic acid sequence.

[00188] In some embodiments, the guide molecule comprises a guide sequence that is designed to have at least one mismatch with the target sequence, such that a heteroduplex formed between the guide sequence and the target sequence comprises a non-pairing C in the guide sequence opposite to the target A for deamination on the target sequence. In some embodiments, aside from this A-C mismatch, the degree of complementarity, when optimally aligned using a suitable alignment algorithm, is about or more than about 50%, 60%, 75%, 80%, 85%, 90%, 95%, 97.5%, 99%, or more.

[00189] In certain embodiments, the guide sequence or spacer length of the guide molecules is from 15 to 50 nt. In certain embodiments, the spacer length of the guide RNA is at least 15 nucleotides. In certain embodiments, the spacer length is from 15 to 17 nt, e.g., 15, 16, or 17 nt, from 17 to 20 nt, e.g., 17, 18, 19, or 20 nt, from 20 to 24 nt, e.g., 20, 21, 22, 23, or 24 nt, from 23 to 25 nt, e.g., 23, 24, or 25 nt, from 24 to 27 nt, e.g., 24, 25, 26, or 27 nt, from 27-30 nt, e.g., 27, 28, 29, or 30 nt, from 30-35 nt, e.g., 30, 31, 32, 33, 34, or 35 nt, or 35 nt or longer. In certain example embodiment, the guide sequence is 15, 16, 17,18, 19, 20, 21, 22, 23, 24, 25, 26, 27, 28, 29, 30, 31, 32, 33, 34, 35, 36, 37, 38, 39 40, 41, 42, 43, 44, 45, 46, 47 48, 49, 50, 51, 52, 53, 54, 55, 56, 57, 58, 59, 60, 61, 62, 63, 64, 65, 66, 67, 68, 69, 70, 71, 72, 73, 74, 75, 76, 77, 78, 79, 80, 81, 82, 83, 84, 85, 86, 87, 88, 89, 90, 91, 92, 93, 94, 95, 96, 97, 98, 99, or 100 nt.

[00190] In some embodiments, the guide sequence is an RNA sequence of between 10 to 50 nt in length, but more particularly of about 20-30 nt advantageously about 20 nt, 23-25 nt or 24 nt. The guide sequence is selected so as to ensure that it hybridizes to the target sequence comprising the adenosine to be deaminated. This is described more in detail below. Selection can encompass further steps which increase efficacy and specificity of deamination.

[00191] In some embodiments, the guide sequence is about 20 nt to about 30 nt long and hybridizes to the target DNA strand to form an almost perfectly matched duplex, except for having a dA-C mismatch at the target adenosine site. Particularly, in some embodiments, the dA-C mismatch is located close to the center of the target sequence (and thus the center of the duplex upon hybridization of the guide sequence to the target sequence), thereby restricting the adenosine deaminase to a narrow editing window (e.g., about 4 bp wide). In some embodiments, the target sequence may comprise more than one target adenosine to be deaminated. In further embodiments the target sequence may further comprise one or more dA-C mismatch 3' to the target adenosine site. In some embodiments, to avoid off-target editing at an unintended Adenine site in the target sequence, the guide sequence can be designed to comprise a non-pairing Guanine at a position corresponding to said unintended Adenine to introduce a dA-G mismatch, which is catalytically unfavorable for certain adenosine deaminases such as ADAR1 and ADAR2. See Wong *et al.*, *RNA* 7:846-858 (2001), which is incorporated herein by reference in its entirety.

[00192] In some embodiments, a Cpf1 guide sequence having a canonical length (e.g., about 24 nt for AsCpf1) is used to form a heteroduplex with the target DNA. In some embodiments, a Cpf1 guide molecule longer than the canonical length (e.g., >24 nt for AsCpf1) is used to form a heteroduplex with the target DNA including outside of the Cpf1-guide RNA-target DNA complex. This can be of interest where deamination of more than one adenine within a given stretch of nucleotides is of interest. In alternative embodiments, it is of interest to maintain the limitation of the canonical guide sequence length. In some embodiments, the guide sequence is designed to introduce a dA-C mismatch outside of the canonical length of Cpf1 guide, which may decrease steric hindrance by Cpf1 and increase the frequency of contact between the adenonsine deaminase and the dA-C mismatch.

[00193] In some embodiments, the position of the mismatched nucleobase (e.g., cytidine) is calculated from where the PAM would be on a DNA target. In some embodiment, the mismatched nucleobase is positioned 12-21 nt from the PAM, or 13-21 nt from the PAM, or 14-21 nt from the PAM, or 14-20 nt from the PAM, or 15-20 nt from the PAM, or 16-20 nt

from the PAM, or 14-19 nt from the PAM, or 15-19 nt from the PAM, or 16-19 nt from the PAM, or 17-19 nt from the PAM, or about 20 nt from the PAM, or about 19 nt from the PAM, or about 18 nt from the PAM, or about 17 nt from the PAM, or about 16 nt from the PAM, or about 15 nt from the PAM, or about 14 nt from the PAM. In a preferred embodiment, the mismatched nucleobase is positioned 17-19 nt or 18 nt from the PAM.

[00194] Mismatch distance is the number of bases between the 3' end of the Cpf1 spacer and the mismatched nucleobase (e.g., cytidine), wherein the mismatched base is included as part of the mismatch distance calculation. In some embodiment, the mismatch distance is 1-10 nt, or 1-9 nt, or 1-8 nt, or 2-8 nt, or 2-7 nt, or 2-6 nt, or 3-8 nt, or 3-7 nt, or 3-6 nt, or 3-5 nt, or about 2 nt, or about 3 nt, or about 4 nt, or about 5 nt, or about 6 nt, or about 7 nt, or about 8 nt. In a preferred embodiment, the mismatch distance is 3-5 nt or 4 nt.

[00195] In some embodiment, the editing window of the Cpf1-ADAR system described herein is 12-21 nt from the PAM, or 13-21 nt from the PAM, or 14-21 nt from the PAM, or 14-20 nt from the PAM, or 15-20 nt from the PAM, or 16-20 nt from the PAM, or 14-19 nt from the PAM, or 15-19 nt from the PAM, or 16-19 nt from the PAM, or 17-19 nt from the PAM, or about 20 nt from the PAM, or about 19 nt from the PAM, or about 18 nt from the PAM, or about 17 nt from the PAM, or about 16 nt from the PAM, or about 15 nt from the PAM, or about 14 nt from the PAM. In some embodiment, the editing window of the Cpf1-ADAR system described herein is 1-10 nt from the 3' end of the Cpf1 spacer, or 1-9 nt from the 3' end of the Cpf1 spacer, or 1-8 nt from the 3' end of the Cpf1 spacer, or 2-8 nt from the 3' end of the Cpf1 spacer, or 2-7 nt from the 3' end of the Cpf1 spacer, or 2-6 nt from the 3' end of the Cpf1 spacer, or 3-8 nt from the 3' end of the Cpf1 spacer, or 3-7 nt from the 3' end of the Cpf1 spacer, or 3-6 nt from the 3' end of the Cpf1 spacer, or 3-5 nt from the 3' end of the Cpf1 spacer, or about 2 nt from the 3' end of the Cpf1 spacer, or about 3 nt from the 3' end of the Cpf1 spacer, or about 4 nt from the 3' end of the Cpf1 spacer, or about 5 nt from the 3' end of the Cpf1 spacer, or about 6 nt from the 3' end of the Cpf1 spacer, or about 7 nt from the 3' end of the Cpf1 spacer, or about 8 nt from the 3' end of the Cpf1 spacer.

[00196] In some embodiments, the sequence of the guide molecule (direct repeat and/or spacer) is selected to reduce the degree secondary structure within the guide molecule. In some embodiments, about or less than about 75%, 50%, 40%, 30%, 25%, 20%, 15%, 10%, 5%, 1%, or fewer of the nucleotides of the nucleic acid-targeting guide RNA participate in self-complementary base pairing when optimally folded. Optimal folding may be determined by any suitable polynucleotide folding algorithm. Some programs are based on

calculating the minimal Gibbs free energy. An example of one such algorithm is mFold, as described by Zuker and Stiegler (Nucleic Acids Res. 9 (1981), 133-148). Another example folding algorithm is the online webserver RNAfold, developed at Institute for Theoretical Chemistry at the University of Vienna, using the centroid structure prediction algorithm (see e.g., A.R. Gruber et al., 2008, Cell 106(1): 23-24; and PA Carr and GM Church, 2009, Nature Biotechnology 27(12): 1151-62).

[00197] In some embodiments, it is of interest to reduce the susceptibility of the guide molecule to RNA cleavage, such as to cleavage by Cpf1. Accordingly, in particular embodiments, the guide molecule is adjusted to avoide cleavage by Cpf1 or other RNA-cleaving enzymes.

[00198] In certain embodiments, the guide molecule comprises non-naturally occurring nucleic acids and/or non-naturally occurring nucleotides and/or nucleotide analogs, and/or chemically modifications. Preferably, these non-naturally occurring nucleic acids and non-naturally occurring nucleotides are located outside the guide sequence. Non-naturally occurring nucleic acids can include, for example, mixtures of naturally and non-naturally occurring nucleotides. Non-naturally occurring nucleotides and/or nucleotide analogs may be modified at the ribose, phosphate, and/or base moiety. In an embodiment of the invention, a guide nucleic acid comprises ribonucleotides and non-ribonucleotides. In one such embodiment, a guide comprises one or more ribonucleotides and one or more deoxyribonucleotides. In an embodiment of the invention, the guide comprises one or more non-naturally occurring nucleotide or nucleotide analog such as a nucleotide with phosphorothioate linkage, a locked nucleic acid (LNA) nucleotides comprising a methylene bridge between the 2′ and 4′ carbons of the ribose ring, or bridged nucleic acids (BNA). Other examples of modified nucleotides include 2'-O-methyl analogs, 2'-deoxy analogs, or 2'-fluoro analogs. Further examples of modified bases include, but are not limited to, 2-aminopurine, 5-bromo-uridine, pseudouridine, inosine, 7-methylguanosine. Examples of guide RNA chemical modifications include, without limitation, incorporation of 2′-O-methyl (M), 2′-O-methyl 3′phosphorothioate (MS), *S*-constrained ethyl(cEt), or 2′-O-methyl 3′thioPACE (MSP) at one or more terminal nucleotides. Such chemically modified guides can comprise increased stability and increased activity as compared to unmodified guides, though on-target vs. off-target specificity is not predictable. (See, Hendel, 2015, Nat Biotechnol. 33(9):985-9, doi: 10.1038/nbt.3290, published online 29 June 2015 Ragdarm et al., 0215, *PNAS*, E7110-E7111; Allerson et al., *J. Med. Chem.* 2005, 48:901-904; Bramsen et al., *Front. Genet.*, 2012, 3:154; Deng et al., *PNAS*, 2015, 112:11870-11875; Sharma et

al., *MedChemComm.*, 2014, 5:1454-1471; Hendel et al., *Nat. Biotechnol.* (2015) 33(9): 985-989; Li et al., *Nature Biomedical Engineering*, 2017, 1, 0066 DOI:10.1038/s41551-017-0066). In some embodiments, the 5' and/or 3' end of a guide RNA is modified by a variety of functional moieties including fluorescent dyes, polyethylene glycol, cholesterol, proteins, or detection tags. (See Kelly et al., 2016, *J. Biotech.* 233:74-83). In certain embodiments, a guide comprises ribonucleotides in a region that binds to a target DNA and one or more deoxyribonucletides and/or nucleotide analogs in a region that binds to Cpf1. In an embodiment of the invention, deoxyribonucleotides and/or nucleotide analogs are incorporated in engineered guide structures, such as, without limitation, stem-loop regions, and the seed region. For Cpf1 guide, in certain embodiments, the modification is not in the 5'-handle of the stem-loop regions. Chemical modification in the 5'-handle of the stem-loop region of a guide may abolish its function (see Li, et al., *Nature Biomedical Engineering*, 2017, 1:0066). In certain embodiments, at least 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20, 21, 22, 23, 24, 25, 26, 27, 28, 29, 30, 35, 40, 45, 50, or 75 nucleotides of a guide is chemically modified. In some embodiments, 3-5 nucleotides at either the 3' or the 5' end of a guide is chemically modified. In some embodiments, only minor modifications are introduced in the seed region, such as 2'-F modifications. In some embodiments, 2'-F modification is introduced at the 3' end of a guide. In certain embodiments, three to five nucleotides at the 5' and/or the 3' end of the guide are chemically modified with 2'-O-methyl (M), 2'-O-methyl 3' phosphorothioate (MS), *S*-constrained ethyl(cEt), or 2'-O-methyl 3' thioPACE (MSP). Such modification can enhance genome editing efficiency (see Hendel et al., *Nat. Biotechnol.* (2015) 33(9): 985-989). In certain embodiments, all of the phosphodiester bonds of a guide are substituted with phosphorothioates (PS) for enhancing levels of gene disruption. In certain embodiments, more than five nucleotides at the 5' and/or the 3' end of the guide are chemically modified with 2'-O-Me, 2'-F or *S*-constrained ethyl(cEt). Such chemically modified guide can mediate enhanced levels of gene disruption (see Ragdarm et al., 0215, *PNAS*, E7110-E7111). In an embodiment of the invention, a guide is modified to comprise a chemical moiety at its 3' and/or 5' end. Such moieties include, but are not limited to amine, azide, alkyne, thio, dibenzocyclooctyne (DBCO), or Rhodamine. In certain embodiment, the chemical moiety is conjugated to the guide by a linker, such as an alkyl chain. In certain embodiments, the chemical moiety of the modified guide can be used to attach the guide to another molecule, such as DNA, RNA, protein, or nanoparticles. Such chemically modified guide can be used to identify or enrich cells

generically edited by a CRISPR system (see Lee et al., *eLife*, 2017, 6:e25312, DOI:10.7554).

[00199] In some embodiments, the guide comprises a modified Cpf1 crRNA, having a 5'-handle and a guide segment further comprising a seed region and a 3'-terminus. In some embodiments, the modified guide can be used with a Cpf1 of any one of Acidaminococcus sp. BV3L6 Cpf1 (AsCpf1); Francisella tularensis subsp. Novicida U112 Cpf1 (FnCpf1); L. bacterium MC2017 Cpf1 (Lb3Cpf1); Butyrivibrio proteoclasticus Cpf1 (BpCpf1); Parcubacteria bacterium GWC2011_GWC2_44_17 Cpf1 (PbCpf1); Peregrinibacteria bacterium GW2011_GWA_33_10 Cpf1 (PeCpf1); Leptospira inadai Cpf1 (LiCpf1); Smithella sp. SC_K08D17 Cpf1 (SsCpf1); L. bacterium MA2020 Cpf1 (Lb2Cpf1); Porphyromonas crevioricanis Cpf1 (PcCpf1); Porphyromonas macacae Cpf1 (PmCpf1); Candidatus Methanoplasma termitum Cpf1 (CMtCpf1); Eubacterium eligens Cpf1 (EeCpf1); Moraxella bovoculi 237 Cpf1 (MbCpf1); Prevotella disiens Cpf1 (PdCpf1); or L. bacterium ND2006 Cpf1 (LbCpf1).

[00200] In some embodiments, the modification to the guide is a chemical modification, an insertion, a deletion or a split. In some embodiments, the chemical modification includes, but is not limited to, incorporation of 2'-O-methyl (M) analogs, 2'-deoxy analogs, 2-thiouridine analogs, N6-methyladenosine analogs, 2'-fluoro analogs, 2-aminopurine, 5-bromo-uridine, pseudouridine (Ψ), N1-methylpseudouridine (me1Ψ), 5-methoxyuridine(5moU), inosine, 7-methylguanosine, 2′-O-methyl 3′phosphorothioate (MS), S-constrained ethyl(cEt), phosphorothioate (PS), or 2′-O-methyl 3′thioPACE (MSP). In some embodiments, the guide comprises one or more of phosphorothioate modifications. In certain embodiments, at least 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20, or 25 nucleotides of the guide are chemically modified. In certain embodiments, one or more nucleotides in the seed region are chemically modified. In certain embodiments, one or more nucleotides in the 3'-terminus are chemically modified. In certain embodiments, none of the nucleotides in the 5'-handle is chemically modified. In some embodiments, the chemical modification in the seed region is a minor modification, such as incorporation of a 2'-fluoro analog. In a specific embodiment, one nucleotide of the seed region is replaced with a 2'-fluoro analog. In some embodiments, 5 to 10 nucleotides in the 3'-terminus are chemically modified. Such chemical modifications at the 3'-terminus of the Cpf1 CrRNA may improve Cpf1 activity (see Li, et al., Nature Biomedical Engineering, 2017, 1:0066). In a specific embodiment, 1, 2, 3, 4, 5, 6, 7, 8, 9 or 10 nucleotides in the 3'-terminus are

replaced with 2'-fluoro analogues. In a specific embodiment, 1, 2, 3, 4, 5, 6, 7, 8, 9 or 10 nucleotides in the 3'-terminus are replaced with 2'- O-methyl (M) analogs.

[00201] In some embodiments, the loop of the 5'-handle of the guide is modified. In some embodiments, the loop of the 5'-handle of the guide is modified to have a deletion, an insertion, a split, or chemical modifications. In certain embodiments, the modified loop comprises 3, 4, or 5 nucleotides. In certain embodiments, the loop comprises the sequence of UCUU, UUUU, UAUU, or UGUU.

[00202] In some embodiments, the guide molecule forms a stem loop with a separate non-covalently linked sequence, which can be DNA or RNA. In particular embodiments, the sequences forming the guide are first synthesized using the standard phosphoramidite synthetic protocol (Herdewijn, P., ed., Methods in Molecular Biology Col 288, Oligonucleotide Synthesis: Methods and Applications, Humana Press, New Jersey (2012)). In some embodiments, these sequences can be functionalized to contain an appropriate functional group for ligation using the standard protocol known in the art (Hermanson, G. T., Bioconjugate Techniques, Academic Press (2013)). Examples of functional groups include, but are not limited to, hydroxyl, amine, carboxylic acid, carboxylic acid halide, carboxylic acid active ester, aldehyde, carbonyl, chlorocarbonyl, imidazolylcarbonyl, hydrozide, semicarbazide, thio semicarbazide, thiol, maleimide, haloalkyl, sufonyl, ally, propargyl, diene, alkyne, and azide. Once this sequence is functionalized, a covalent chemical bond or linkage can be formed between this sequence and the direct repeat sequence. Examples of chemical bonds include, but are not limited to, those based on carbamates, ethers, esters, amides, imines, amidines, aminotrizines, hydrozone, disulfides, thioethers, thioesters, phosphorothioates, phosphorodithioates, sulfonamides, sulfonates, fulfones, sulfoxides, ureas, thioureas, hydrazide, oxime, triazole, photolabile linkages, C-C bond forming groups such as Diels-Alder cyclo-addition pairs or ring-closing metathesis pairs, and Michael reaction pairs.

[00203] In some embodiments, these stem-loop forming sequences can be chemically synthesized. In some embodiments, the chemical synthesis uses automated, solid-phase oligonucleotide synthesis machines with 2'-acetoxyethyl orthoester (2'-ACE) (Scaringe et al., J. Am. Chem. Soc. (1998) 120: 11820-11821; Scaringe, Methods Enzymol. (2000) 317: 3-18) or 2'-thionocarbamate (2'-TC) chemistry (Dellinger et al., J. Am. Chem. Soc. (2011) 133: 11540-11546; Hendel et al., Nat. Biotechnol. (2015) 33:985-989).

[00204] In certain embodiments, the guide molecule (capable of guiding Cpf1 to a target locus) comprises (1) a guide sequence capable of hybridizing to a target locus and (2) a tracr

mate or direct repeat sequence whereby the direct repeat sequence is located upstream (i.e., 5') from the guide sequence. In a particular embodiment the seed sequence (i.e. the sequence essential critical for recognition and/or hybridization to the sequence at the target locus) of the Cpf1 guide sequence is approximately within the first 10 nucleotides of the guide sequence. In particular embodiments, the Cpf1 is FnCpf1 and the seed sequence is approximately within the first 5 nt on the 5' end of the guide sequence.

[00205] In a particular embodiment the guide molecule comprises a guide sequence linked to a direct repeat sequence, wherein the direct repeat sequence comprises one or more stem loops or optimized secondary structures. In particular embodiments, the direct repeat has a minimum length of 16 nts and a single stem loop. In further embodiments the direct repeat has a length longer than 16 nts, preferrably more than 17 nts, and has more than one stem loops or optimized secondary structures. In particular embodiments the guide molecule comprises or consists of the guide sequence linked to all or part of the natural direct repeat sequence. A typical Type V Cpf1 guide molecule comprises (in 3' to 5' direction): a guide sequence a first complimentary stretch (the "repeat"), a loop (which is typically 4 or 5 nucleotides long), a second complimentary stretch (the "anti-repeat" being complimentary to the repeat), and a poly A (often poly U in RNA) tail (terminator). In certain embodiments, the direct repeat sequence retains its natural architercture and forms a single stem loop. In particular embodiments, certain aspects of the guide architecture can be modified, for example by addition, subtraction, or substitution of features, whereas certain other aspects of guide architecture are maintained. Preferred locations for engineered guide molecule modifications, including but not limited to insertions, deletions, and substitutions include guide termini and regions of the guide molecule that are exposed when complexed with the Cpf1 protein and/or target, for example the stemloop of the direct repeat sequence.

[00206] In particular embodiments, the stem comprises at least about 4bp comprising complementary X and Y sequences, although stems of more, e.g., 5, 6, 7, 8, 9, 10, 11 or 12 or fewer, e.g., 3, 2, base pairs are also contemplated. Thus, for example X2-10 and Y2-10 (wherein X and Y represent any complementary set of nucleotides) may be contemplated. In one aspect, the stem made of the X and Y nucleotides, together with the loop will form a complete hairpin in the overall secondary structure; and, this may be advantageous and the amount of base pairs can be any amount that forms a complete hairpin. In one aspect, any complementary X:Y basepairing sequence (e.g., as to length) is tolerated, so long as the secondary structure of the entire guide molecule is preserved. In one aspect, the loop that connects the stem made of X:Y basepairs can be any sequence of the same length (e.g., 4

or 5 nucleotides) or longer that does not interrupt the overall secondary structure of the guide molecule. In one aspect, the stemloop can further comprise, e.g. an MS2 aptamer. In one aspect, the stem comprises about 5-7bp comprising complementary X and Y sequences, although stems of more or fewer basepairs are also contemplated. In one aspect, non-Watson Crick basepairing is contemplated, where such pairing otherwise generally preserves the architecture of the stemloop at that position.

[00207] In particular embodiments the natural hairpin or stemloop structure of the guide molecule is extended or replaced by an extended stemloop. It has been demonstrated that extension of the stem can enhance the assembly of the guide molecule with the CRISPR-Cas proten (Chen et al. Cell. (2013); 155(7): 1479–1491). In particular embodiments the stem of the stemloop is extended by at least 1, 2, 3, 4, 5 or more complementary basepairs (i.e. corresponding to the addition of 2,4, 6, 8, 10 or more nucleotides in the guide molecule). In particular embodiments these are located at the end of the stem, adjacent to the loop of the stemloop.

[00208] In particular embodiments, the susceptibility of the guide molecule to RNAses or to decreased expression can be reduced by slight modifications of the sequence of the guide molecule which do not affect its function. For instance, in particular embodiments, premature termination of transcription, such as premature transcription of U6 Pol-III, can be removed by modifying a putative Pol-III terminator (4 consecutive U's) in the guide molecules sequence. Where such sequence modification is required in the stemloop of the guide molecule, it is preferably ensured by a basepair flip.

[00209] In a preferred embodiment the direct repeat may be modified to comprise one or more protein-binding RNA aptamers. In a particular embodiment, one or more aptamers may be included such as part of optimized secondary structure. Such aptamers may be capable of binding a bacteriophage coat protein as detailed further herein.

[00210] In some embodiments, the guide molecule forms a duplex with a target DNA strand comprising at least one target adenosine residues to be edited. Upon hybridization of the guide RNA molecule to the target DNA strand, the adenosine deaminase binds to the duplex and catalyzes deamination of one or more target adenosine residues comprised within the DNA-RNA duplex.

[00211] A guide sequence, and hence a nucleic acid-targeting guide RNA may be selected to target any target nucleic acid sequence. The target sequence may be DNA. The target sequence may be genomic DNA. The target sequence may be mitochondrial DNA.

[00212] In certain embodiments, the target sequence should be associated with a PAM (protospacer adjacent motif) or PFS (protospacer flanking sequence or site); that is, a short sequence recognized by the CRISPR complex. Depending on the nature of the CRISPR-Cas protein, the target sequence should be selected such that its complementary sequence in the DNA duplex (also referred to herein as the non-target sequence) is upstream or downstream of the PAM. In the embodiments of the present invention where the CRISPR-Cas protein is a Cpf1 protein, the compelementary sequence of the target sequence in a is downstream or 3' of the PAM. The precise sequence and length requirements for the PAM differ depending on the Cpf1 protein used, but PAMs are typically 2-5 base pair sequences adjacent the protospacer (that is, the target sequence). Examples of the natural PAM sequences for different Cpf1 orthologues are provided herein below and the skilled person will be able to identify further PAM sequences for use with a given Cpf1 protein.

[00213] Further, engineering of the PAM Interacting (PI) domain may allow programing of PAM specificity, improve target site recognition fidelity, and increase the versatility of the CRISPR-Cas protein, for example as described for Cas9 in Kleinstiver BP et al. Engineered CRISPR-Cas9 nucleases with altered PAM specificities. Nature. 2015 Jul 23;523(7561):481-5. doi: 10.1038/nature14592. As further detailed herein, the skilled person will understand that Cpf1 proteins may be modified analogously.

[00214] In particular embodiments, the guide sequence is selected in order to ensure optimal efficiency of the deaminase on the adenine to be deaminated. The position of the adenine in the target strand relative to the cleavage site of the Cpf1 nickase may be taken into account. In particular embodiments it is of interest to ensure that the nickase will act in the vicinity of the adenine to be deaminated, on the non-target strand. For instance, in particular embodiments, the Cpf1 nickase cuts the non-targeting strand 17 nucleotides downstream of the PAM (e.g. AsCpf1, LbCpf1) or 18 nucleotides downstream of the PAM (e.g. FnCpf1), and it can be of interest to design the guide that the cytosine which is to correspond to the adenine to be deaminated is located in the guide sequence within 10 bp upstream or downstream of the nickase cleavage site in the sequence of the corresponding non-target strand.

[00215] In particular embodiment, the guide is an escorted guide. By "escorted" is meant that the Cpf1 CRISPR-Cas system or complex or guide is delivered to a selected time or place within a cell, so that activity of the Cpf1 CRISPR-Cas system or complex or guide is spatially or temporally controlled. For example, the activity and destination of the Cpf1 CRISPR-Cas system or complex or guide may be controlled by an escort RNA aptamer

sequence that has binding affinity for an aptamer ligand, such as a cell surface protein or other localized cellular component. Alternatively, the escort aptamer may for example be responsive to an aptamer effector on or in the cell, such as a transient effector, such as an external energy source that is applied to the cell at a particular time.

[00216] The escorted Cpf1 CRISPR-Cas systems or complexes have a guide molecule with a functional structure designed to improve guide molecule structure, architecture, stability, genetic expression, or any combination thereof. Such a structure can include an aptamer.

[00217] Aptamers are biomolecules that can be designed or selected to bind tightly to other ligands, for example using a technique called systematic evolution of ligands by exponential enrichment (SELEX; Tuerk C, Gold L: "Systematic evolution of ligands by exponential enrichment: RNA ligands to bacteriophage T4 DNA polymerase." Science 1990, 249:505-510). Nucleic acid aptamers can for example be selected from pools of random-sequence oligonucleotides, with high binding affinities and specificities for a wide range of biomedically relevant targets, suggesting a wide range of therapeutic utilities for aptamers (Keefe, Anthony D., Supriya Pai, and Andrew Ellington. "Aptamers as therapeutics." Nature Reviews Drug Discovery 9.7 (2010): 537-550). These characteristics also suggest a wide range of uses for aptamers as drug delivery vehicles (Levy-Nissenbaum, Etgar, et al. "Nanotechnology and aptamers: applications in drug delivery." Trends in biotechnology 26.8 (2008): 442-449; and, Hicke BJ, Stephens AW. "Escort aptamers: a delivery service for diagnosis and therapy." J Clin Invest 2000, 106:923-928.). Aptamers may also be constructed that function as molecular switches, responding to a que by changing properties, such as RNA aptamers that bind fluorophores to mimic the activity of green flourescent protein (Paige, Jeremy S., Karen Y. Wu, and Samie R. Jaffrey. "RNA mimics of green fluorescent protein." Science 333.6042 (2011): 642-646). It has also been suggested that aptamers may be used as components of targeted siRNA therapeutic delivery systems, for example targeting cell surface proteins (Zhou, Jiehua, and John J. Rossi. "Aptamer-targeted cell-specific RNA interference." Silence 1.1 (2010): 4).

[00218] Accordingly, in particular embodiments, the guide molecule is modified, e.g., by one or more aptamer(s) designed to improve guide molecule delivery, including delivery across the cellular membrane, to intracellular compartments, or into the nucleus. Such a structure can include, either in addition to the one or more aptamer(s) or without such one or more aptamer(s), moiety(ies) so as to render the guide molecule deliverable, inducible or responsive to a selected effector. The invention accordingly comprehends an guide

molecule that responds to normal or pathological physiological conditions, including without limitation pH, hypoxia, O_2 concentration, temperature, protein concentration, enzymatic concentration, lipid structure, light exposure, mechanical disruption (e.g. ultrasound waves), magnetic fields, electric fields, or electromagnetic radiation.

[00219] Light responsiveness of an inducible system may be achieved via the activation and binding of cryptochrome-2 and CIB1. Blue light stimulation induces an activating conformational change in cryptochrome-2, resulting in recruitment of its binding partner CIB1. This binding is fast and reversible, achieving saturation in <15 sec following pulsed stimulation and returning to baseline <15 min after the end of stimulation. These rapid binding kinetics result in a system temporally bound only by the speed of transcription/translation and transcript/protein degradation, rather than uptake and clearance of inducing agents. Crytochrome-2 activation is also highly sensitive, allowing for the use of low light intensity stimulation and mitigating the risks of phototoxicity. Further, in a context such as the intact mammalian brain, variable light intensity may be used to control the size of a stimulated region, allowing for greater precision than vector delivery alone may offer.

[00220] The invention contemplates energy sources such as electromagnetic radiation, sound energy or thermal energy to induce the guide. Advantageously, the electromagnetic radiation is a component of visible light. In a preferred embodiment, the light is a blue light with a wavelength of about 450 to about 495 nm. In an especially preferred embodiment, the wavelength is about 488 nm. In another preferred embodiment, the light stimulation is via pulses. The light power may range from about 0-9 mW/cm^2. In a preferred embodiment, a stimulation paradigm of as low as 0.25 sec every 15 sec should result in maximal activation.

[00221] The chemical or energy sensitive guide may undergo a conformational change upon induction by the binding of a chemical source or by the energy allowing it act as a guide and have the Cpf1 CRISPR-Cas system or complex function. The invention can involve applying the chemical source or energy so as to have the guide function and the Cpf1 CRISPR-Cas system or complex function; and optionally further determining that the expression of the genomic locus is altered.

[00222] There are several different designs of this chemical inducible system: 1. ABI-PYL based system inducible by Abscisic Acid (ABA) (see, e.g., http://stke.sciencemag.org/cgi/content/abstract/sigtrans;4/164/rs2), 2. FKBP-FRB based system inducible by rapamycin (or related chemicals based on rapamycin) (see, e.g.,

http://www.nature.com/nmeth/journal/v2/n6/full/nmeth763.html), 3. GID1-GAI based system inducible by Gibberellin (GA) (see, e.g., http://www.nature.com/nchembio/journal/v8/n5/full/nchembio.922.html).

[00223] A chemical inducible system can be an estrogen receptor (ER) based system inducible by 4-hydroxytamoxifen (4OHT) (see, e.g., http://www.pnas.org/content/104/3/1027.abstract). A mutated ligand-binding domain of the estrogen receptor called ERT2 translocates into the nucleus of cells upon binding of 4-hydroxytamoxifen. In further embodiments of the invention any naturally occurring or engineered derivative of any nuclear receptor, thyroid hormone receptor, retinoic acid receptor, estrogren receptor, estrogen-related receptor, glucocorticoid receptor, progesterone receptor, androgen receptor may be used in inducible systems analogous to the ER based inducible system.

[00224] Another inducible system is based on the design using Transient receptor potential (TRP) ion channel based system inducible by energy, heat or radio-wave (see, e.g., http://www.sciencemag.org/content/336/6081/604). These TRP family proteins respond to different stimuli, including light and heat. When this protein is activated by light or heat, the ion channel will open and allow the entering of ions such as calcium into the plasma membrane. This influx of ions will bind to intracellular ion interacting partners linked to a polypeptide including the guide and the other components of the Cpf1 CRISPR-Cas complex or system, and the binding will induce the change of sub-cellular localization of the polypeptide, leading to the entire polypeptide entering the nucleus of cells. Once inside the nucleus, the guide protein and the other components of the Cpf1 CRISPR-Cas complex will be active and modulating target gene expression in cells.

[00225] While light activation may be an advantageous embodiment, sometimes it may be disadvantageous especially for in vivo applications in which the light may not penetrate the skin or other organs. In this instance, other methods of energy activation are contemplated, in particular, electric field energy and/or ultrasound which have a similar effect.

[00226] Electric field energy is preferably administered substantially as described in the art, using one or more electric pulses of from about 1 Volt/cm to about 10 kVolts/cm under *in vivo* conditions. Instead of or in addition to the pulses, the electric field may be delivered in a continuous manner. The electric pulse may be applied for between 1 μs and 500 milliseconds, preferably between 1 μs and 100 milliseconds. The electric field may be applied continuously or in a pulsed manner for 5 about minutes.

[00227] As used herein, 'electric field energy' is the electrical energy to which a cell is exposed. Preferably the electric field has a strength of from about 1 Volt/cm to about 10 kVolts/cm or more under *in vivo* conditions (see WO97/49450).

[00228] As used herein, the term "electric field" includes one or more pulses at variable capacitance and voltage and including exponential and/or square wave and/or modulated wave and/or modulated square wave forms. References to electric fields and electricity should be taken to include reference the presence of an electric potential difference in the environment of a cell. Such an environment may be set up by way of static electricity, alternating current (AC), direct current (DC), etc, as known in the art. The electric field may be uniform, non-uniform or otherwise, and may vary in strength and/or direction in a time dependent manner.

[00229] Single or multiple applications of electric field, as well as single or multiple applications of ultrasound are also possible, in any order and in any combination. The ultrasound and/or the electric field may be delivered as single or multiple continuous applications, or as pulses (pulsatile delivery).

[00230] Electroporation has been used in both *in vitro* and *in vivo* procedures to introduce foreign material into living cells. With *in vitro* applications, a sample of live cells is first mixed with the agent of interest and placed between electrodes such as parallel plates. Then, the electrodes apply an electrical field to the cell/implant mixture. Examples of systems that perform *in vitro* electroporation include the Electro Cell Manipulator ECM600 product, and the Electro Square Porator T820, both made by the BTX Division of Genetronics, Inc (see U.S. Pat. No 5,869,326).

[00231] The known electroporation techniques (both *in vitro* and *in vivo*) function by applying a brief high voltage pulse to electrodes positioned around the treatment region. The electric field generated between the electrodes causes the cell membranes to temporarily become porous, whereupon molecules of the agent of interest enter the cells. In known electroporation applications, this electric field comprises a single square wave pulse on the order of 1000 V/cm, of about 100 .mu.s duration. Such a pulse may be generated, for example, in known applications of the Electro Square Porator T820.

[00232] Preferably, the electric field has a strength of from about 1 V/cm to about 10 kV/cm under *in vitro* conditions. Thus, the electric field may have a strength of 1 V/cm, 2 V/cm, 3 V/cm, 4 V/cm, 5 V/cm, 6 V/cm, 7 V/cm, 8 V/cm, 9 V/cm, 10 V/cm, 20 V/cm, 50 V/cm, 100 V/cm, 200 V/cm, 300 V/cm, 400 V/cm, 500 V/cm, 600 V/cm, 700 V/cm, 800 V/cm, 900 V/cm, 1 kV/cm, 2 kV/cm, 5 kV/cm, 10 kV/cm, 20 kV/cm, 50 kV/cm or more.

More preferably from about 0.5 kV/cm to about 4.0 kV/cm under *in vitro* conditions. Preferably the electric field has a strength of from about 1 V/cm to about 10 kV/cm under *in vivo* conditions. However, the electric field strengths may be lowered where the number of pulses delivered to the target site are increased. Thus, pulsatile delivery of electric fields at lower field strengths is envisaged.

[00233] Preferably the application of the electric field is in the form of multiple pulses such as double pulses of the same strength and capacitance or sequential pulses of varying strength and/or capacitance. As used herein, the term "pulse" includes one or more electric pulses at variable capacitance and voltage and including exponential and/or square wave and/or modulated wave/square wave forms.

[00234] Preferably the electric pulse is delivered as a waveform selected from an exponential wave form, a square wave form, a modulated wave form and a modulated square wave form.

[00235] A preferred embodiment employs direct current at low voltage. Thus, Applicants disclose the use of an electric field which is applied to the cell, tissue or tissue mass at a field strength of between 1V/cm and 20V/cm, for a period of 100 milliseconds or more, preferably 15 minutes or more.

[00236] Ultrasound is advantageously administered at a power level of from about 0.05 W/cm^2 to about 100 W/cm^2. Diagnostic or therapeutic ultrasound may be used, or combinations thereof.

[00237] As used herein, the term "ultrasound" refers to a form of energy which consists of mechanical vibrations the frequencies of which are so high they are above the range of human hearing. Lower frequency limit of the ultrasonic spectrum may generally be taken as about 20 kHz. Most diagnostic applications of ultrasound employ frequencies in the range 1 and 15 MHz' (From Ultrasonics in Clinical Diagnosis, P. N. T. Wells, ed., 2nd. Edition, Publ. Churchill Livingstone [Edinburgh, London & NY, 1977]).

[00238] Ultrasound has been used in both diagnostic and therapeutic applications. When used as a diagnostic tool ("diagnostic ultrasound"), ultrasound is typically used in an energy density range of up to about 100 mW/cm^2 (FDA recommendation), although energy densities of up to 750 mW/cm^2 have been used. In physiotherapy, ultrasound is typically used as an energy source in a range up to about 3 to 4 W/cm^2 (WHO recommendation). In other therapeutic applications, higher intensities of ultrasound may be employed, for example, HIFU at 100 W/cm up to 1 kW/cm^2 (or even higher) for short periods of time. The

term "ultrasound" as used in this specification is intended to encompass diagnostic, therapeutic and focused ultrasound.

[00239] Focused ultrasound (FUS) allows thermal energy to be delivered without an invasive probe (see Morocz et al 1998 Journal of Magnetic Resonance Imaging Vol.8, No. 1, pp.136-142. Another form of focused ultrasound is high intensity focused ultrasound (HIFU) which is reviewed by Moussatov et al in Ultrasonics (1998) Vol.36, No.8, pp.893-900 and TranHuuHue et al in Acustica (1997) Vol.83, No.6, pp.1103-1106.

[00240] Preferably, a combination of diagnostic ultrasound and a therapeutic ultrasound is employed. This combination is not intended to be limiting, however, and the skilled reader will appreciate that any variety of combinations of ultrasound may be used. Additionally, the energy density, frequency of ultrasound, and period of exposure may be varied.

[00241] Preferably the exposure to an ultrasound energy source is at a power density of from about 0.05 to about 100 Wcm^{-2}. Even more preferably, the exposure to an ultrasound energy source is at a power density of from about 1 to about 15 Wcm^{-2}.

[00242] Preferably the exposure to an ultrasound energy source is at a frequency of from about 0.015 to about 10.0 MHz. More preferably the exposure to an ultrasound energy source is at a frequency of from about 0.02 to about 5.0 MHz or about 6.0 MHz. Most preferably, the ultrasound is applied at a frequency of 3 MHz.

[00243] Preferably the exposure is for periods of from about 10 milliseconds to about 60 minutes. Preferably the exposure is for periods of from about 1 second to about 5 minutes. More preferably, the ultrasound is applied for about 2 minutes. Depending on the particular target cell to be disrupted, however, the exposure may be for a longer duration, for example, for 15 minutes.

[00244] Advantageously, the target tissue is exposed to an ultrasound energy source at an acoustic power density of from about 0.05 Wcm^{-2} to about 10 Wcm^{-2} with a frequency ranging from about 0.015 to about 10 MHz (see WO 98/52609). However, alternatives are also possible, for example, exposure to an ultrasound energy source at an acoustic power density of above 100 Wcm^{-2}, but for reduced periods of time, for example, 1000 Wcm^{-2} for periods in the millisecond range or less.

[00245] Preferably the application of the ultrasound is in the form of multiple pulses; thus, both continuous wave and pulsed wave (pulsatile delivery of ultrasound) may be employed in any combination. For example, continuous wave ultrasound may be applied, followed by pulsed wave ultrasound, or vice versa. This may be repeated any number of

times, in any order and combination. The pulsed wave ultrasound may be applied against a background of continuous wave ultrasound, and any number of pulses may be used in any number of groups.

[00246] Preferably, the ultrasound may comprise pulsed wave ultrasound. In a highly preferred embodiment, the ultrasound is applied at a power density of 0.7 Wcm-2 or 1.25 Wcm-2 as a continuous wave. Higher power densities may be employed if pulsed wave ultrasound is used.

[00247] Use of ultrasound is advantageous as, like light, it may be focused accurately on a target. Moreover, ultrasound is advantageous as it may be focused more deeply into tissues unlike light. It is therefore better suited to whole-tissue penetration (such as but not limited to a lobe of the liver) or whole organ (such as but not limited to the entire liver or an entire muscle, such as the heart) therapy. Another important advantage is that ultrasound is a non-invasive stimulus which is used in a wide variety of diagnostic and therapeutic applications. By way of example, ultrasound is well known in medical imaging techniques and, additionally, in orthopedic therapy. Furthermore, instruments suitable for the application of ultrasound to a subject vertebrate are widely available and their use is well known in the art.

[00248] In particular embodiments, the guide molecule is modified by a secondary structure to increase the specificity of the CRISPR-Cas system and the secondary structure can protect against exonuclease activity and allow for 5' additions to the guide sequence also referred to herein as a protected guide molecule.

[00249] In one aspect, the invention provides for hybridizing a "protector RNA" to a sequence of the guide molecule, wherein the "protector RNA" is an RNA strand complementary to the 3' end of the guide molecule to thereby generate a partially double-stranded guide RNA. In an embodiment of the invention, protecting mismatched bases (i.e. the bases of the guide molecule which do not form part of the guide sequence) with a perfectly complementary protector sequence decreases the likelihood of target DNA binding to the mismatched basepairs at the 3' end. In particular embodiments of the invention, additional sequences comprising an extented length may also be present within the guide molecule such that the guide comprises a protector sequence within the guide molecule. This "protector sequence" ensures that the guide molecule comprises a "protected sequence" in addition to an "exposed sequence" (comprising the part of the guide sequence hybridizing to the target sequence). In particular embodiments, the guide molecule is modified by the presence of the protector guide to comprise a secondary structure such as a

hairpin. Advantageously there are three or four to thirty or more, e.g., about 10 or more, contiguous base pairs having complementarity to the protected sequence, the guide sequence or both. It is advantageous that the protected portion does not impede thermodynamics of the CRISPR-Cas system interacting with its target. By providing such an extension including a partially double stranded guide moleucle, the guide molecule is considered protected and results in improved specific binding of the CRISPR-Cas complex, while maintaining specific activity.

[00250] In particular embodiments, use is made of a truncated guide (tru-guide), i.e. a guide molecule which comprises a guide sequence which is truncated in length with respect to the canonical guide sequence length. As described by Nowak et al. (Nucleic Acids Res (2016) 44 (20): 9555-9564), such guides may allow catalytically active CRISPR-Cas enzyme to bind its target without cleaving the target DNA. In particular embodiments, a truncated guide is used which allows the binding of the target but retains only nickase activity of the CRISPR-Cas enzyme.

[00251] *Crispr-Cas Enzyme*

[00252] In certain embodiments of the methods provided herein the CRISPR-Cas protein has reduced or no catalytic activity. Where the CRISPR-Cas protein is a Cpf1 protein, the mutations may include but are not limited to one or more mutations in the catalytic RuvC-like domain, such as D908A or E993A with reference to the positions in AsCpf1.

[00253] In some embodiments, a CRISPR-Cas protein is considered to substantially lack all DNA cleavage activity when the DNA cleavage activity of the mutated enzyme is about no more than 25%, 10%, 5%, 1%, 0.1%, 0.01%, or less of the DNA cleavage activity of the non-mutated form of the enzyme; an example can be when the DNA cleavage activity of the mutated form is nil or negligible as compared with the non-mutated form. In these embodiments, the CRISPR-Cas protein is used as a generic DNA binding protein. The mutations may be artificially introduced mutations or gain- or loss-of-function mutations.

[00254] In addition to the mutations described above, the CRISPR-Cas protein may be additionally modified. As used herein, the term "modified" with regard to a CRISPR-Cas protein generally refers to a CRISPR-Cas protein having one or more modifications or mutations (including point mutations, truncations, insertions, deletions, chimeras, fusion proteins, etc.) compared to the wild type Cas protein from which it is derived. By derived is meant that the derived enzyme is largely based, in the sense of having a high degree of sequence homology with, a wildtype enzyme, but that it has been mutated (modified) in some way as known in the art or as described herein.

[00255] The additional modifications of the CRISPR-Cas protein may or may not cause an altered functionality. By means of example, and in particular with reference to CRISPR-Cas protein, modifications which do not result in an altered functionality include for instance codon optimization for expression into a particular host, or providing the nuclease with a particular marker (e.g. for visualization). Modifications with may result in altered functionality may also include mutations, including point mutations, insertions, deletions, truncations (including split nucleases), etc.. Fusion proteins may without limitation include for instance fusions with heterologous domains or functional domains (e.g. localization signals, catalytic domains, etc.). In certain embodiments, various different modifications may be combined (e.g. a mutated nuclease which is catalytically inactive and which further is fused to a functional domain, such as for instance to induce DNA methylation or another nucleic acid modification, such as including without limitation a break (e.g. by a different nuclease (domain)), a mutation, a deletion, an insertion, a replacement, a ligation, a digestion, a break or a recombination). As used herein, "altered functionality" includes without limitation an altered specificity (e.g. altered target recognition, increased (e.g. "enhanced" Cas proteins) or decreased specificity, or altered PAM recognition), altered activity (e.g. increased or decreased catalytic activity, including catalytically inactive nucleases or nickases), and/or altered stability (e.g. fusions with destalilization domains). Suitable heterologous domains include without limitation a nuclease, a ligase, a repair protein, a methyltransferase, (viral) integrase, a recombinase, a transposase, an argonaute, a cytidine deaminase, a retron, a group II intron, a phosphatase, a phosphorylase, a sulpfurylase, a kinase, a polymerase, an exonuclease, etc.. Examples of all these modifications are known in the art. It will be understood that a "modified" nuclease as referred to herein, and in particular a "modified" Cas or "modified" CRISPR-Cas system or complex preferably still has the capacity to interact with or bind to the polynucleic acid (e.g. in complex with theguide molecule). Such modified Cas protein can be combined with the deaminase protein or active domain thereof as described herein.

[00256] In certain embodiments, CRISPR-Cas protein may comprise one or more modifications resulting in enhanced activity and/or specificity, such as including mutating residues that stabilize the targeted or non-targeted strand (e.g. eCas9; "Rationally engineered Cas9 nucleases with improved specificity", Slaymaker et al. (2016), Science, 351(6268):84-88, incorporated herewith in its entirety by reference). In certain embodiments, the altered or modified activity of the engineered CRISPR protein comprises increased targeting efficiency or decreased off-target binding. In certain embodiments, the

altered activity of the engineered CRISPR protein comprises modified cleavage activity. In certain embodiments, the altered activity comprises increased cleavage activity as to the target polynucleotide loci. In certain embodiments, the altered activity comprises decreased cleavage activity as to the target polynucleotide loci. In certain embodiments, the altered activity comprises decreased cleavage activity as to off-target polynucleotide loci. In certain embodiments, the altered or modified activity of the modified nuclease comprises altered helicase kinetics. In certain embodiments, the modified nuclease comprises a modification that alters association of the protein with the nucleic acid molecule comprising RNA (in the case of a Cas protein), or a strand of the target polynucleotide loci, or a strand of off-target polynucleotide loci. In an aspect of the invention, the engineered CRISPR protein comprises a modification that alters formation of the CRISPR complex. In certain embodiments, the altered activity comprises increased cleavage activity as to off-target polynucleotide loci. Accordingly, in certain embodiments, there is increased specificity for target polynucleotide loci as compared to off-target polynucleotide loci. In other embodiments, there is reduced specificity for target polynucleotide loci as compared to off-target polynucleotide loci. In certain embodiments, the mutations result in decreased off-target effects (e.g. cleavage or binding properties, activity, or kinetics), such as in case for Cas proteins for instance resulting in a lower tolerance for mismatches between target and guide RNA. Other mutations may lead to increased off-target effects (e.g. cleavage or binding properties, activity, or kinetics). Other mutations may lead to increased or decreased on-target effects (e.g. cleavage or binding properties, activity, or kinetics). In certain embodiments, the mutations result in altered (e.g. increased or decreased) helicase activity, association or formation of the functional nuclease complex (e.g. CRISPR-Cas complex). In certain embodiments, as described above, the mutations result in an altered PAM recognition, i.e. a different PAM may be (in addition or in the alternative) be recognized, compared to the unmodified Cas protein. Particularly preferred mutations include positively charged residues and/or (evolutionary) conserved residues, such as conserved positively charged residues, in order to enhance specificity. In certain embodiments, such residues may be mutated to uncharged residues, such as alanine.

[00257] Base Excision Repair Inhibitor

[00258] In some embodiments, the AD-functionalized CRISPR system further comprises a base excision repair (BER) inhibitor. Without wishing to be bound by any particular theory, cellular DNA-repair response to the presence of I:T pairing may be responsible for a decrease in nucleobase editing efficiency in cells. Alkyladenine DNA glycosylase (also

known as DNA-3-methyladenine glycosylase, 3-alkyladenine DNA glycosylase, or N-methylpurine DNA glycosylase) catalyzes removal of hypoxanthine from DNA in cells, which may initiate base excision repair, with reversion of the I:T pair to a A:T pair as outcome.

[00259] In some embodiments, the BER inhibitor is an inhibitor of alkyladenine DNA glycosylase. In some embodiments, the BER inhibitor is an inhibitor of human alkyladenine DNA glycosylase. In some embodiments, the BER inhibitor is a polypeptide inhibitor. In some embodiments, the BER inhibitor is a protein that binds hypoxanthine. In some embodiments, the BER inhibitor is a protein that binds hypoxanthine in DNA. In some embodiments, the BER inhibitor is a catalytically inactive alkyladenine DNA glycosylase protein or binding domain thereof. In some embodiments, the BER inhibitor is a catalytically inactive alkyladenine DNA glycosylase protein or binding domain thereof that does not excise hypoxanthine from the DNA. Other proteins that are capable of inhibiting (e.g., sterically blocking) an alkyladenine DNA glycosylase base-excision repair enzyme are within the scope of this disclosure. Additionally, any proteins that block or inhibit base-excision repair as also within the scope of this disclosure.

[00260] Without wishing to be bound by any particular theory, base excision repair may be inhibited by molecules that bind the edited strand, block the edited base, inhibit alkyladenine DNA glycosylase, inhibit base excision repair, protect the edited base, and/or promote fixing of the non-edited strand. It is believed that the use of the BER inhibitor described herein can increase the editing efficiency of an adenosine deaminase that is capable of catalyzing a A to I change.

[00261] Accordingly, in the first design of the AD-functionalized CRISPR system discussed above, the CRISPR-Cas protein or the adenosine deaminase can be fused to or linked to a BER inhibitor (e.g., an inhibitor of alkyladenine DNA glycosylase). In some embodiments, the BER inhibitor can be comprised in one of the following structures (nCpf1=Cpf1 nickase; dCpf1=dead Cpf1):

[AD]-[optional linker]-[nCpf1/dCpf1]-[optional linker]-[BER inhibitor];
[AD]-[optional linker]-[BER inhibitor]-[optional linker]-[nCpf1/dCpf1];
[BER inhibitor]-[optional linker]-[AD]-[optional linker]-[nCpf1/dCpf1];
[BER inhibitor]-[optional linker]-[nCpf1/dCpf1]-[optional linker]-[AD];
[nCpf1/dCpf1]-[optional linker]-[AD]-[optional linker]-[BER inhibitor];
[nCpf1/dCpf1]-[optional linker]-[BER inhibitor]-[optional linker]-[AD].

[00262] Similarly, in the second design of the AD-functionalized CRISPR system discussed above, the CRISPR-Cas protein, the adenosine deaminase, or the adaptor protein can be fused to or linked to a BER inhibitor (e.g., an inhibitor of alkyladenine DNA glycosylase). In some embodiments, the BER inhibitor can be comprised in one of the following structures (nCpf1=Cpf1 nickase; dCpf1=dead Cpf1):

[nCpf1/dCpf1]-[optional linker]-[BER inhibitor];

[BER inhibitor]-[optional linker]-[nCpf1/dCpf1];

[AD]-[optional linker]-[Adaptor]-[optional linker]-[BER inhibitor];

[AD]-[optional linker]-[BER inhibitor]-[optional linker]-[Adaptor];

[BER inhibitor]-[optional linker]-[AD]-[optional linker]-[Adaptor];

[BER inhibitor]-[optional linker]-[Adaptor]-[optional linker]-[AD];

[Adaptor]-[optional linker]-[AD]-[optional linker]-[BER inhibitor];

[Adaptor]-[optional linker]-[BER inhibitor]-[optional linker]-[AD].

[00263] In the third design of the AD-functionalized CRISPR system discussed above, the BER inhibitor can be inserted into an internal loop or unstructured region of a CRISPR-Cas protein.

[00264] **Targeting to the Nucleus**

[00265] In some embodiments, the methods of the present invention relate to modifying an Adenine in a target locus of interest, whereby the target locus is within a cell. In order to improve targeting of the CRISPR-Cas protein and/or the adenosine deaminase protein or catalytic domain thereof used in the methods of the present invention to the nucleus, it may be advantageous to provide one or both of these components with one or more nuclear localization sequences (NLSs).

[00266] In preferred embodiments, the NLSs used in the context of the present invention are heterologous to the proteins. Non-limiting examples of NLSs include an NLS sequence derived from: the NLS of the SV40 virus large T-antigen, having the amino acid sequence PKKKRKV (SEQ ID NO: 37) or PKKKRKVEAS (SEQ ID NO: 38); the NLS from nucleoplasmin (e.g., the nucleoplasmin bipartite NLS with the sequence KRPAATKKAGQAKKKK (SEQ ID NO: 39)); the c-myc NLS having the amino acid sequence PAAKRVKLD (SEQ ID NO: 40) or RQRRNELKRSP (SEQ ID NO: 41); the hRNPA1 M9 NLS having the sequence NQSSNFGPMKGGNFGGRSSGPYGGGGQYFAKPRNQGGY (SEQ ID NO: 42); the sequence RMRIZFKNKGKDTAELRRRRVEVSVELRKAKKDEQILKRRNV (SEQ ID NO: 43) of the IBB domain from importin-alpha; the sequences VSRKRPRP (SEQ ID NO:

44) and PPKKARED (SEQ ID NO: 45) of the myoma T protein; the sequence PQPKKKPL (SEQ ID NO: 46) of human p53; the sequence SALIKKKKKMAP (SEQ ID NO: 47) of mouse c-abl IV; the sequences DRLRR (SEQ ID NO: 48) and PKQKKRK (SEQ ID NO: 49) of the influenza virus NS1; the sequence RKLKKKIKKL (SEQ ID NO: 50) of the Hepatitis virus delta antigen; the sequence REKKKFLKRR (SEQ ID NO: 51) of the mouse Mx1 protein; the sequence KRKGDEVDGVDEVAKKKSKK (SEQ ID NO: 52) of the human poly(ADP-ribose) polymerase; and the sequence RKCLQAGMNLEARKTKK (SEQ ID NO: 53) of the steroid hormone receptors (human) glucocorticoid. In general, the one or more NLSs are of sufficient strength to drive accumulation of the DNA-targeting Cas protein in a detectable amount in the nucleus of a eukaryotic cell. In general, strength of nuclear localization activity may derive from the number of NLSs in the CRISPR-Cas protein, the particular NLS(s) used, or a combination of these factors. Detection of accumulation in the nucleus may be performed by any suitable technique. For example, a detectable marker may be fused to the nucleic acid-targeting protein, such that location within a cell may be visualized, such as in combination with a means for detecting the location of the nucleus (e.g., a stain specific for the nucleus such as DAPI). Cell nuclei may also be isolated from cells, the contents of which may then be analyzed by any suitable process for detecting protein, such as immunohistochemistry, Western blot, or enzyme activity assay. Accumulation in the nucleus may also be determined indirectly, such as by an assay for the effect of nucleic acid-targeting complex formation (e.g., assay for deaminase activity) at the target sequence, or assay for altered gene expression activity affected by DNA-targeting complex formation and/or DNA-targeting), as compared to a control not exposed to the CRISPR-Cas protein and deaminase protein, or exposed to a CRISPR-Cas and/or deaminase protein lacking the one or more NLSs.

[00267] The CRISPR-Cas and/or adenosine deaminase proteins may be provided with 1 or more, such as with, 2, 3, 4, 5, 6, 7, 8, 9, 10, or more heterologous NLSs. In some embodiments, the proteins comprises about or more than about 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, or more NLSs at or near the amino-terminus, about or more than about 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, or more NLSs at or near the carboxy-terminus, or a combination of these (e.g., zero or at least one or more NLS at the amino-terminus and zero or at one or more NLS at the carboxy terminus). When more than one NLS is present, each may be selected independently of the others, such that a single NLS may be present in more than one copy and/or in combination with one or more other NLSs present in one or more copies. In some embodiments, an NLS is considered near the N- or C-terminus when the nearest amino acid

of the NLS is within about 1, 2, 3, 4, 5, 10, 15, 20, 25, 30, 40, 50, or more amino acids along the polypeptide chain from the N- or C-terminus. In preferred embodiments of the CRISPR-Cas proteins, an NLS attached to the C-terminal of the protein.

[00268] In certain embodiments of the methods provided herein, the CRISPR-Cas protein and the deaminase protein are delivered to the cell or expressed within the cell as separate proteins. In these embodiments, each of the CRISPR-Cas and deaminase protein can be provided with one or more NLSs as described herein. In certain embodiments, the CRISPR-Cas and deaminase proteins are delivered to the cell or expressed with the cell as a fusion protein. In these embodiments one or both of the CRISPR-Cas and deaminase protein is provided with one or more NLSs. Where the adenosine deaminase is fused to an adaptor protein (such as MS2) as described above, the one or more NLS can be provided on the adaptor protein, provided that this does not interfere with aptamer binding. In particular embodiments, the one or more NLS sequences may also function as linker sequences between the adenosine deaminase and the CRISPR-Cas protein.

[00269] In certain embodiments, guides of the invention comprise specific binding sites (e.g. aptamers) for adapter proteins, which may be linked to or fused to an adenosine deaminase or catalytic domain thereof. When such a guides forms a CRISPR complex (i.e. CRISPR-Cas protein binding to guide and target) the adapter proteins bind and, the adenosine deaminase or catalytic domain thereof associated with the adapter protein is positioned in a spatial orientation which is advantageous for the attributed function to be effective.

[00270] The skilled person will understand that modifications to the guide which allow for binding of the adapter + adenosine deaminase, but not proper positioning of the adapter + adenosine deaminase (e.g. due to steric hindrance within the three dimensional structure of the CRISPR complex) are modifications which are not intended. The one or more modified guide may be modified at the tetra loop, the stem loop 1, stem loop 2, or stem loop 3, as described herein, preferably at either the tetra loop or stem loop 2, and most preferably at both the tetra loop and stem loop 2.

[00271] Use of orthogonal catalytically inactive CRISPR-Cas proteins

[00272] In particular embodiments, the Cpf1 nickase is used in combination with an orthogonal catalytically inactive CRISPR-Cas protein to increase efficiency of said Cpf1 nickase (as described in Chen et al. 2017, Nature Communications 8:14958;

doi:10.1038/ncomms14958). More particularly, the orthogonal catalytically inactive CRISPR-Cas protein is characterized by a different PAM recognition site than the Cpf1 nickase used in the AD-functionalized CRISPR system and the corresponding guide sequence is selected to bind to a target sequence proximal to that of the Cpf1 nickase of the AD-functionalized CRISPR system. The orthogonal catalytically inactive CRISPR-Cas protein as used in the context of the present invention does not form part of the AD-functionalized CRISPR system but merely functions to increase the efficiency of said Cpf1 nickase and is used in combination with a standard guide molecule as described in the art for said CRISPR-Cas protein. In particular embodiments, said orthogonal catalytically inactive CRISPR-Cas protein is a dead CRISPR-Cas protein, i.e. comprising one or more mutations which abolishes the nuclease activity of said CRISPR-Cas protein. In particular embodiments, the catalytically inactive orthogonal CRISPR-Cas protein is provided with two or more guide molecules which are capable of hybridizing to target sequences which are proximal to the target sequence of the Cpf1 nickase. In particular embodiments, at least two guide molecules are used to target said catalytically inactive CRISPR-Cas protein, of which at least one guide molecule is capable of hybridizing to a target sequence 5" of the target sequence of the Cpf1 nickase and at least one guide molecule is capable of hybridizing to a target sequence 3' of the target sequence of the Cpf1 nickase of the AD-functionalized CRISPR system, whereby said one or more target sequences may be on the same or the opposite DNA strand as the target sequence of the Cpf1 nickase. In particular embodiments, the guide sequences for the one or more guide molecules of the orthogonal catalytically inactive CRISPR-Cas protein are selected such that the target sequences are proximal to that of the guide molecule for the targeting of the AD-functionalized CRISPR, i.e. for the targeting of the Cpf1 nickase. In particular embodiments, the one or more target sequences of the orthogonal catalytically inactive CRISPR-Cas enzyme are each separated from the target sequence of the Cpf1 nickase by more than 5 but less than 450 basepairs. Optimal distances between the target sequences of the guides for use with the orthogonal catalytically inactive CRISPR-Cas protein and the target sequence of the AD-functionalized CRISPR system can be determined by the skilled person. In particular embodiments, the orthogonal CRISPR-Cas protein is a Class II, type II CRISPR protein. In particular embodiments, the orthogonal CRISPR-Cas protein is a Class II, type V CRISPR protein. In particular embodiments, the catalytically inactive orthogonal CRISPR-Cas protein In particular embodiments, the catalytically inactive orthogonal CRISPR-Cas protein has been modified to alter its PAM specificity as described elsewhere herein. In particular

embodiments, the Cpf1 protein nickase is a nickase which, by itself has limited activity in human cells, but which, in combination with an inactive orthogonal CRISPR-Cas protein and one or more corresponding proximal guides ensures the required nickase activity.

[00273] CRISPR Development and Use

[00274] The present invention may be further illustrated and extended based on aspects of CRISPR-Cas development and use as set forth in the following articles and particularly as relates to delivery of a CRISPR protein complex and uses of an RNA guided endonuclease in cells and organisms:

- Multiplex genome engineering using CRISPR-Cas systems. Cong, L., Ran, F.A., Cox, D., Lin, S., Barretto, R., Habib, N., Hsu, P.D., Wu, X., Jiang, W., Marraffini, L.A., & Zhang, F. Science Feb 15;339(6121):819-23 (2013);
- RNA-guided editing of bacterial genomes using CRISPR-Cas systems. Jiang W., Bikard D., Cox D., Zhang F, Marraffini LA. Nat Biotechnol Mar;31(3):233-9 (2013);
- One-Step Generation of Mice Carrying Mutations in Multiple Genes by CRISPR-Cas-Mediated Genome Engineering. Wang H., Yang H., Shivalila CS., Dawlaty MM., Cheng AW., Zhang F., Jaenisch R. Cell May 9;153(4):910-8 (2013);
- Optical control of mammalian endogenous transcription and epigenetic states. Konermann S, Brigham MD, Trevino AE, Hsu PD, Heidenreich M, Cong L, Platt RJ, Scott DA, Church GM, Zhang F. Nature. Aug 22;500(7463):472-6. doi: 10.1038/Nature12466. Epub 2013 Aug 23 (2013);
- Double Nicking by RNA-Guided CRISPR Cas9 for Enhanced Genome Editing Specificity. Ran, FA., Hsu, PD., Lin, CY., Gootenberg, JS., Konermann, S., Trevino, AE., Scott, DA., Inoue, A., Matoba, S., Zhang, Y., & Zhang, F. Cell Aug 28. pii: S0092-8674(13)01015-5 (2013-A);
- DNA targeting specificity of RNA-guided Cas9 nucleases. Hsu, P., Scott, D., Weinstein, J., Ran, FA., Konermann, S., Agarwala, V., Li, Y., Fine, E., Wu, X., Shalem, O., Cradick, TJ., Marraffini, LA., Bao, G., & Zhang, F. Nat Biotechnol doi:10.1038/nbt.2647 (2013);
- Genome engineering using the CRISPR-Cas9 system. Ran, FA., Hsu, PD., Wright, J., Agarwala, V., Scott, DA., Zhang, F. Nature Protocols Nov;8(11):2281-308 (2013-B);

- Genome-Scale CRISPR-Cas9 Knockout Screening in Human Cells. Shalem, O., Sanjana, NE., Hartenian, E., Shi, X., Scott, DA., Mikkelson, T., Heckl, D., Ebert, BL., Root, DE., Doench, JG., Zhang, F. Science Dec 12. (2013);
- Crystal structure of cas9 in complex with guide RNA and target DNA. Nishimasu, H., Ran, FA., Hsu, PD., Konermann, S., Shehata, SI., Dohmae, N., Ishitani, R., Zhang, F., Nureki, O. Cell Feb 27, 156(5):935-49 (2014);
- Genome-wide binding of the CRISPR endonuclease Cas9 in mammalian cells. Wu X., Scott DA., Kriz AJ., Chiu AC., Hsu PD., Dadon DB., Cheng AW., Trevino AE., Konermann S., Chen S., Jaenisch R., Zhang F., Sharp PA. Nat Biotechnol. Apr 20. doi: 10.1038/nbt.2889 (2014);
- CRISPR-Cas9 Knockin Mice for Genome Editing and Cancer Modeling. Platt RJ, Chen S, Zhou Y, Yim MJ, Swiech L, Kempton HR, Dahlman JE, Parnas O, Eisenhaure TM, Jovanovic M, Graham DB, Jhunjhunwala S, Heidenreich M, Xavier RJ, Langer R, Anderson DG, Hacohen N, Regev A, Feng G, Sharp PA, Zhang F. Cell 159(2): 440-455 DOI: 10.1016/j.cell.2014.09.014(2014);
- Development and Applications of CRISPR-Cas9 for Genome Engineering, Hsu PD, Lander ES, Zhang F., Cell. Jun 5;157(6):1262-78 (2014).
- Genetic screens in human cells using the CRISPR-Cas9 system, Wang T, Wei JJ, Sabatini DM, Lander ES., Science. January 3; 343(6166): 80–84. doi:10.1126/science.1246981 (2014);
- Rational design of highly active sgRNAs for CRISPR-Cas9-mediated gene inactivation, Doench JG, Hartenian E, Graham DB, Tothova Z, Hegde M, Smith I, Sullender M, Ebert BL, Xavier RJ, Root DE., (published online 3 September 2014) Nat Biotechnol. Dec;32(12):1262-7 (2014);
- *In vivo* interrogation of gene function in the mammalian brain using CRISPR-Cas9, Swiech L, Heidenreich M, Banerjee A, Habib N, Li Y, Trombetta J, Sur M, Zhang F., (published online 19 October 2014) Nat Biotechnol. Jan;33(1):102-6 (2015);
- Genome-scale transcriptional activation by an engineered CRISPR-Cas9 complex, Konermann S, Brigham MD, Trevino AE, Joung J, Abudayyeh OO, Barcena C, Hsu PD, Habib N, Gootenberg JS, Nishimasu H, Nureki O, Zhang F., Nature. Jan 29;517(7536):583-8 (2015).
- A split-Cas9 architecture for inducible genome editing and transcription modulation, Zetsche B, Volz SE, Zhang F., (published online 02 February 2015) Nat Biotechnol. Feb;33(2):139-42 (2015);

- Genome-wide CRISPR Screen in a Mouse Model of Tumor Growth and Metastasis, Chen S, Sanjana NE, Zheng K, Shalem O, Lee K, Shi X, Scott DA, Song J, Pan JQ, Weissleder R, Lee H, Zhang F, Sharp PA. Cell 160, 1246–1260, March 12, 2015 (multiplex screen in mouse), and
- In *vivo* genome editing using *Staphylococcus aureus* Cas9, Ran FA, Cong L, Yan WX, Scott DA, Gootenberg JS, Kriz AJ, Zetsche B, Shalem O, Wu X, Makarova KS, Koonin EV, Sharp PA, Zhang F., (published online 01 April 2015), Nature. Apr 9;520(7546):186-91 (2015).
- Shalem et al., "High-throughput functional genomics using CRISPR-Cas9," Nature Reviews Genetics 16, 299-311 (May 2015).
- Xu et al., "Sequence determinants of improved CRISPR sgRNA design," Genome Research 25, 1147-1157 (August 2015).
- Parnas et al., "A Genome-wide CRISPR Screen in Primary Immune Cells to Dissect Regulatory Networks," Cell 162, 675-686 (July 30, 2015).
- Ramanan et al., CRISPR-Cas9 cleavage of viral DNA efficiently suppresses hepatitis B virus," Scientific Reports 5:10833. doi: 10.1038/srep10833 (June 2, 2015)
- Nishimasu et al., Crystal Structure of Staphylococcus aureus Cas9," Cell 162, 1113-1126 (Aug. 27, 2015)
- BCL11A enhancer dissection by Cas9-mediated in situ saturating mutagenesis, Canver et al., Nature 527(7577):192-7 (Nov. 12, 2015) doi: 10.1038/nature15521. Epub 2015 Sep 16.
- *Cpf1 Is a Single RNA-Guided Endonuclease of a Class 2 CRISPR-Cas System*, Zetsche et al., Cell 163, 759-71 (Sep 25, 2015).
- *Discovery and Functional Characterization of Diverse Class 2 CRISPR-Cas Systems*, Shmakov et al., Molecular Cell, 60(3), 385–397 doi: 10.1016/j.molcel.2015.10.008 Epub October 22, 2015.
- *Rationally engineered Cas9 nucleases with improved specificity*, Slaymaker et al., Science 2016 Jan 1 351(6268): 84-88 doi: 10.1126/science.aad5227. Epub 2015 Dec 1.
- Gao *et al*, "Engineered Cpf1 Enzymes with Altered PAM Specificities," bioRxiv 091611; doi: http://dx.doi.org/10.1101/091611 (Dec. 4, 2016).

each of which is incorporated herein by reference, may be considered in the practice of the instant invention, and discussed briefly below:

- Cong *et al.* engineered type II CRISPR-Cas systems for use in eukaryotic cells based on both *Streptococcus thermophilus* Cas9 and also *Streptococcus pyogenes* Cas9 and demonstrated that Cas9 nucleases can be directed by short RNAs to induce precise cleavage of DNA in human and mouse cells. Their study further showed that Cas9 as converted into a nicking enzyme can be used to facilitate homology-directed repair in eukaryotic cells with minimal mutagenic activity. Additionally, their study demonstrated that multiple guide sequences can be encoded into a single CRISPR array to enable simultaneous editing of several at endogenous genomic loci sites within the mammalian genome, demonstrating easy programmability and wide applicability of the RNA-guided nuclease technology. This ability to use RNA to program sequence specific DNA cleavage in cells defined a new class of genome engineering tools. These studies further showed that other CRISPR loci are likely to be transplantable into mammalian cells and can also mediate mammalian genome cleavage. Importantly, it can be envisaged that several aspects of the CRISPR-Cas system can be further improved to increase its efficiency and versatility.
- Jiang *et al.* used the clustered, regularly interspaced, short palindromic repeats (CRISPR)–associated Cas9 endonuclease complexed with dual-RNAs to introduce precise mutations in the genomes of *Streptococcus pneumoniae* and *Escherichia coli.* The approach relied on dual-RNA:Cas9-directed cleavage at the targeted genomic site to kill unmutated cells and circumvents the need for selectable markers or counter-selection systems. The study reported reprogramming dual-RNA:Cas9 specificity by changing the sequence of short CRISPR RNA (crRNA) to make single- and multinucleotide changes carried on editing templates. The study showed that simultaneous use of two crRNAs enabled multiplex mutagenesis. Furthermore, when the approach was used in combination with recombineering, in *S. pneumoniae*, nearly 100% of cells that were recovered using the described approach contained the desired mutation, and in *E. coli,* 65% that were recovered contained the mutation.
- Wang *et al.* (2013) used the CRISPR-Cas system for the one-step generation of mice carrying mutations in multiple genes which were traditionally generated in multiple steps by sequential recombination in embryonic stem cells and/or time-consuming intercrossing of mice with a single mutation. The CRISPR-Cas system will greatly accelerate the *in vivo* study of functionally redundant genes and of epistatic gene interactions.

- Konermann *et al.* (2013) addressed the need in the art for versatile and robust technologies that enable optical and chemical modulation of DNA-binding domains based CRISPR Cas9 enzyme and also Transcriptional Activator Like Effectors
- Ran *et al.* (2013-A) described an approach that combined a Cas9 nickase mutant with paired guide RNAs to introduce targeted double-strand breaks. This addresses the issue of the Cas9 nuclease from the microbial CRISPR-Cas system being targeted to specific genomic loci by a guide sequence, which can tolerate certain mismatches to the DNA target and thereby promote undesired off-target mutagenesis. Because individual nicks in the genome are repaired with high fidelity, simultaneous nicking *via* appropriately offset guide RNAs is required for double-stranded breaks and extends the number of specifically recognized bases for target cleavage. The authors demonstrated that using paired nicking can reduce off-target activity by 50- to 1,500-fold in cell lines and to facilitate gene knockout in mouse zygotes without sacrificing on-target cleavage efficiency. This versatile strategy enables a wide variety of genome editing applications that require high specificity.
- Hsu *et al.* (2013) characterized SpCas9 targeting specificity in human cells to inform the selection of target sites and avoid off-target effects. The study evaluated >700 guide RNA variants and SpCas9-induced indel mutation levels at >100 predicted genomic off-target loci in 293T and 293FT cells. The authors that SpCas9 tolerates mismatches between guide RNA and target DNA at different positions in a sequence-dependent manner, sensitive to the number, position and distribution of mismatches. The authors further showed that SpCas9-mediated cleavage is unaffected by DNA methylation and that the dosage of SpCas9 and guide RNA can be titrated to minimize off-target modification. Additionally, to facilitate mammalian genome engineering applications, the authors reported providing a web-based software tool to guide the selection and validation of target sequences as well as off-target analyses.
- Ran *et al.* (2013-B) described a set of tools for Cas9-mediated genome editing *via* non-homologous end joining (NHEJ) or homology-directed repair (HDR) in mammalian cells, as well as generation of modified cell lines for downstream functional studies. To minimize off-target cleavage, the authors further described a double-nicking strategy using the Cas9 nickase mutant with paired guide RNAs. The protocol provided by the authors experimentally derived guidelines for the selection of target sites, evaluation of cleavage efficiency and analysis of off-target activity.

The studies showed that beginning with target design, gene modifications can be achieved within as little as 1–2 weeks, and modified clonal cell lines can be derived within 2–3 weeks.

- Shalem *et al.* described a new way to interrogate gene function on a genome-wide scale. Their studies showed that delivery of a genome-scale CRISPR-Cas9 knockout (GeCKO) library targeted 18,080 genes with 64,751 unique guide sequences enabled both negative and positive selection screening in human cells. First, the authors showed use of the GeCKO library to identify genes essential for cell viability in cancer and pluripotent stem cells. Next, in a melanoma model, the authors screened for genes whose loss is involved in resistance to vemurafenib, a therapeutic that inhibits mutant protein kinase BRAF. Their studies showed that the highest-ranking candidates included previously validated genes NF1 and MED12 as well as novel hits NF2, CUL3, TADA2B, and TADA1. The authors observed a high level of consistency between independent guide RNAs targeting the same gene and a high rate of hit confirmation, and thus demonstrated the promise of genome-scale screening with Cas9.
- Nishimasu *et al.* reported the crystal structure of *Streptococcus pyogenes* Cas9 in complex with sgRNA and its target DNA at 2.5 A° resolution. The structure revealed a bilobed architecture composed of target recognition and nuclease lobes, accommodating the sgRNA:DNA heteroduplex in a positively charged groove at their interface. Whereas the recognition lobe is essential for binding sgRNA and DNA, the nuclease lobe contains the HNH and RuvC nuclease domains, which are properly positioned for cleavage of the complementary and non-complementary strands of the target DNA, respectively. The nuclease lobe also contains a carboxyl-terminal domain responsible for the interaction with the protospacer adjacent motif (PAM). This high-resolution structure and accompanying functional analyses have revealed the molecular mechanism of RNA-guided DNA targeting by Cas9, thus paving the way for the rational design of new, versatile genome-editing technologies.
- Wu *et al.* mapped genome-wide binding sites of a catalytically inactive Cas9 (dCas9) from *Streptococcus pyogenes* loaded with single guide RNAs (sgRNAs) in mouse embryonic stem cells (mESCs). The authors showed that each of the four sgRNAs tested targets dCas9 to between tens and thousands of genomic sites, frequently characterized by a 5-nucleotide seed region in the sgRNA and an NGG

protospacer adjacent motif (PAM). Chromatin inaccessibility decreases dCas9 binding to other sites with matching seed sequences; thus 70% of off-target sites are associated with genes. The authors showed that targeted sequencing of 295 dCas9 binding sites in mESCs transfected with catalytically active Cas9 identified only one site mutated above background levels. The authors proposed a two-state model for Cas9 binding and cleavage, in which a seed match triggers binding but extensive pairing with target DNA is required for cleavage.

- Platt *et al.* established a Cre-dependent Cas9 knockin mouse. The authors demonstrated *in vivo* as well as *ex vivo* genome editing using adeno-associated virus (AAV)-, lentivirus-, or particle-mediated delivery of guide RNA in neurons, immune cells, and endothelial cells.
- Hsu *et al.* (2014) is a review article that discusses generally CRISPR-Cas9 history from yogurt to genome editing, including genetic screening of cells.
- Wang *et al.* (2014) relates to a pooled, loss-of-function genetic screening approach suitable for both positive and negative selection that uses a genome-scale lentiviral single guide RNA (sgRNA) library.
- Doench *et al.* created a pool of sgRNAs, tiling across all possible target sites of a panel of six endogenous mouse and three endogenous human genes and quantitatively assessed their ability to produce null alleles of their target gene by antibody staining and flow cytometry. The authors showed that optimization of the PAM improved activity and also provided an on-line tool for designing sgRNAs.
- Swiech *et al.* demonstrate that AAV-mediated SpCas9 genome editing can enable reverse genetic studies of gene function in the brain.
- Konermann *et al.* (2015) discusses the ability to attach multiple effector domains, e.g., transcriptional activator, functional and epigenomic regulators at appropriate positions on the guide such as stem or tetraloop with and without linkers.
- Zetsche *et al.* demonstrates that the Cas9 enzyme can be split into two and hence the assembly of Cas9 for activation can be controlled.
- Chen *et al.* relates to multiplex screening by demonstrating that a genome-wide *in vivo* CRISPR-Cas9 screen in mice reveals genes regulating lung metastasis.
- Ran *et al.* (2015) relates to SaCas9 and its ability to edit genomes and demonstrates that one cannot extrapolate from biochemical assays.
- Shalem *et al.* (2015) described ways in which catalytically inactive Cas9 (dCas9) fusions are used to synthetically repress (CRISPRi) or activate (CRISPRa)

expression, showing. advances using Cas9 for genome-scale screens, including arrayed and pooled screens, knockout approaches that inactivate genomic loci and strategies that modulate transcriptional activity.

- Xu *et al.* (2015) assessed the DNA sequence features that contribute to single guide RNA (sgRNA) efficiency in CRISPR-based screens. The authors explored efficiency of CRISPR-Cas9 knockout and nucleotide preference at the cleavage site. The authors also found that the sequence preference for CRISPRi/a is substantially different from that for CRISPR-Cas9 knockout.
- Parnas *et al.* (2015) introduced genome-wide pooled CRISPR-Cas9 libraries into dendritic cells (DCs) to identify genes that control the induction of tumor necrosis factor (Tnf) by bacterial lipopolysaccharide (LPS). Known regulators of Tlr4 signaling and previously unknown candidates were identified and classified into three functional modules with distinct effects on the canonical responses to LPS.
- Ramanan *et al* (2015) demonstrated cleavage of viral episomal DNA (cccDNA) in infected cells. The HBV genome exists in the nuclei of infected hepatocytes as a 3.2kb double-stranded episomal DNA species called covalently closed circular DNA (cccDNA), which is a key component in the HBV life cycle whose replication is not inhibited by current therapies. The authors showed that sgRNAs specifically targeting highly conserved regions of HBV robustly suppresses viral replication and depleted cccDNA.
- Nishimasu *et al.* (2015) reported the crystal structures of SaCas9 in complex with a single guide RNA (sgRNA) and its double-stranded DNA targets, containing the 5'-TTGAAT-3' PAM and the 5'-TTGGGT-3' PAM. A structural comparison of SaCas9 with SpCas9 highlighted both structural conservation and divergence, explaining their distinct PAM specificities and orthologous sgRNA recognition.
- Canver *et al.* (2015) demonstrated a CRISPR-Cas9-based functional investigation of non-coding genomic elements. The authors we developed pooled CRISPR-Cas9 guide RNA libraries to perform *in situ* saturating mutagenesis of the human and mouse BCL11A enhancers which revealed critical features of the enhancers.
- Zetsche et al. (2015) reported characterization of Cpf1, a class 2 CRISPR nuclease from Francisella novicida U112 having features distinct from Cas9. Cpf1 is a single RNA-guided endonuclease lacking tracrRNA, utilizes a T-rich protospacer-adjacent motif, and cleaves DNA via a staggered DNA double-stranded break.

- Shmakov et al. (2015) reported three distinct Class 2 CRISPR-Cas systems. Two system CRISPR enzymes (C2c1 and C2c3) contain RuvC-like endonuclease domains distantly related to Cpf1. Unlike Cpf1, C2c1 depends on both crRNA and tracrRNA for DNA cleavage. The third enzyme (C2c2) contains two predicted HEPN RNase domains and is tracrRNA independent.
- Slaymaker et al (2016) reported the use of structure-guided protein engineering to improve the specificity of Streptococcus pyogenes Cas9 (SpCas9). The authors developed "enhanced specificity" SpCas9 (eSpCas9) variants which maintained robust on-target cleavage with reduced off-target effects.

[00275] The methods and tools provided herein are exemplified for Cpf1, a type II nuclease that does not make use of tracrRNA. Orthologs of Cpf1 have been identified in different bacterial species as described herein. Further type II nucleases with similar properties can be identified using methods described in the art (Shmakov et al. 2015, 60:385–397; Abudayeh et al. 2016, Science, 5;353(6299)) . In particular embodiments, such methods for identifying novel CRISPR effector proteins may comprise the steps of selecting sequences from the database encoding a seed which identifies the presence of a CRISPR Cas locus, identifying loci located within 10 kb of the seed comprising Open Reading Frames (ORFs) in the selected sequences, selecting therefrom loci comprising ORFs of which only a single ORF encodes a novel CRISPR effector having greater than 700 amino acids and no more than 90% homology to a known CRISPR effector. In particular embodiments, the seed is a protein that is common to the CRISPR-Cas system, such as Cas1. In further embodiments, the CRISPR array is used as a seed to identify new effector proteins.

[00276] The effectiveness of the present invention has been demonstrated. Preassembled recombinant CRISPR-Cpf1 complexes comprising Cpf1 and crRNA may be transfected, for example by electroporation, resulting in high mutation rates and absence of detectable off-target mutations. Hur, J.K. et al, Targeted mutagenesis in mice by electroporation of Cpf1 ribonucleoproteins, Nat Biotechnol. 2016 Jun 6. doi: 10.1038/nbt.3596. Genome-wide analyses shows that Cpf1 is highly specific. By one measure, *in vitro* cleavage sites determined for Cpf1 in human HEK293T cells were significantly fewer that for SpCas9. Kim, D. et al., Genome-wide analysis reveals specificities of Cpf1 endonucleases in human cells, Nat Biotechnol. 2016 Jun 6. doi: 10.1038/nbt.3609. An efficient multiplexed system employing Cpf1 has been demonstrated in Drosophila employing gRNAs processed from an

array containing inventing tRNAs. Port, F. et al, Expansion of the CRISPR toolbox in an animal with tRNA-flanked Cas9 and Cpf1 gRNAs. doi: http://dx.doi.org/10.1101/046417.

[00277] Also, "Dimeric CRISPR RNA-guided FokI nucleases for highly specific genome editing", Shengdar Q. Tsai, Nicolas Wyvekens, Cyd Khayter, Jennifer A. Foden, Vishal Thapar, Deepak Reyon, Mathew J. Goodwin, Martin J. Aryee, J. Keith Joung Nature Biotechnology 32(6): 569-77 (2014), relates to dimeric RNA-guided FokI Nucleases that recognize extended sequences and can edit endogenous genes with high efficiencies in human cells.

[00278] With respect to general information on CRISPR/Cas Systems, components thereof, and delivery of such components, including methods, materials, delivery vehicles, vectors, particles, and making and using thereof, including as to amounts and formulations, as well as CRISPR-Cas-expressing eukaryotic cells, CRISPR-Cas expressing eukaryotes, such as a mouse, reference is made to: US Patents Nos. 8,999,641, 8,993,233, 8,697,359, 8,771,945, 8,795,965, 8,865,406, 8,871,445, 8,889,356, 8,889,418, 8,895,308, 8,906,616, 8,932,814, and 8,945,839; US Patent Publications US 2014-0310830 (US App. Ser. No. 14/105,031), US 2014-0287938 A1 (U.S. App. Ser. No. 14/213,991), US 2014-0273234 A1 (U.S. App. Ser. No. 14/293,674), US2014-0273232 A1 (U.S. App. Ser. No. 14/290,575), US 2014-0273231 (U.S. App. Ser. No. 14/259,420), US 2014-0256046 A1 (U.S. App. Ser. No. 14/226,274), US 2014-0248702 A1 (U.S. App. Ser. No. 14/258,458), US 2014-0242700 A1 (U.S. App. Ser. No. 14/222,930), US 2014-0242699 A1 (U.S. App. Ser. No. 14/183,512), US 2014-0242664 A1 (U.S. App. Ser. No. 14/104,990), US 2014-0234972 A1 (U.S. App. Ser. No. 14/183,471), US 2014-0227787 A1 (U.S. App. Ser. No. 14/256,912), US 2014-0189896 A1 (U.S. App. Ser. No. 14/105,035), US 2014-0186958 (U.S. App. Ser. No. 14/105,017), US 2014-0186919 A1 (U.S. App. Ser. No. 14/104,977), US 2014-0186843 A1 (U.S. App. Ser. No. 14/104,900), US 2014-0179770 A1 (U.S. App. Ser. No. 14/104,837) and US 2014-0179006 A1 (U.S. App. Ser. No. 14/183,486), US 2014-0170753 (US App Ser No 14/183,429); US 2015-0184139 (U.S. App. Ser. No. 14/324,960); 14/054,414 European Patent Applications EP 2 771 468 (EP13818570.7), EP 2 764 103 (EP13824232.6), and EP 2 784 162 (EP14170383.5); and PCT Patent Publications WO2014/093661 (PCT/US2013/074743), WO2014/093694 (PCT/US2013/074790), WO2014/093595 (PCT/US2013/074611), WO2014/093718 (PCT/US2013/074825), WO2014/093709 (PCT/US2013/074812), WO2014/093622 (PCT/US2013/074667), WO2014/093635 (PCT/US2013/074691), WO2014/093655 (PCT/US2013/074736), WO2014/093712 (PCT/US2013/074819), WO2014/093701 (PCT/US2013/074800),

WO2014/018423 (PCT/US2013/051418), WO2014/204723 (PCT/US2014/041790), WO2014/204724 (PCT/US2014/041800), WO2014/204725 (PCT/US2014/041803), WO2014/204726 (PCT/US2014/041804), WO2014/204727 (PCT/US2014/041806), WO2014/204728 (PCT/US2014/041808), WO2014/204729 (PCT/US2014/041809), WO2015/089351 (PCT/US2014/069897), WO2015/089354 (PCT/US2014/069902), WO2015/089364 (PCT/US2014/069925), WO2015/089427 (PCT/US2014/070068), WO2015/089462 (PCT/US2014/070127), WO2015/089419 (PCT/US2014/070057), WO2015/089465 (PCT/US2014/070135), WO2015/089486 (PCT/US2014/070175), WO2015/058052 (PCT/US2014/061077), WO2015/070083 (PCT/US2014/064663), WO2015/089354 (PCT/US2014/069902), WO2015/089351 (PCT/US2014/069897), WO2015/089364 (PCT/US2014/069925), WO2015/089427 (PCT/US2014/070068), WO2015/089473 (PCT/US2014/070152), WO2015/089486 (PCT/US2014/070175), WO2016/049258 (PCT/US2015/051830), WO2016/094867 (PCT/US2015/065385), WO2016/094872 (PCT/US2015/065393), WO2016/094874 (PCT/US2015/065396), WO2016/106244 (PCT/US2015/067177).

[00279] Mention is also made of US application 62/180,709, 17-Jun-15, PROTECTED GUIDE RNAS (PGRNAS); US application 62/091,455, filed, 12-Dec-14, PROTECTED GUIDE RNAS (PGRNAS); US application 62/096,708, 24-Dec-14, PROTECTED GUIDE RNAS (PGRNAS); US applications 62/091,462, 12-Dec-14, 62/096,324, 23-Dec-14, 62/180,681, 17-Jun-2015, and 62/237,496, 5-Oct-2015, DEAD GUIDES FOR CRISPR TRANSCRIPTION FACTORS; US application 62/091,456, 12-Dec-14 and 62/180,692, 17-Jun-2015, ESCORTED AND FUNCTIONALIZED GUIDES FOR CRISPR-CAS SYSTEMS; US application 62/091,461, 12-Dec-14, DELIVERY, USE AND THERAPEUTIC APPLICATIONS OF THE CRISPR-CAS SYSTEMS AND COMPOSITIONS FOR GENOME EDITING AS TO HEMATOPOETIC STEM CELLS (HSCs); US application 62/094,903, 19-Dec-14, UNBIASED IDENTIFICATION OF DOUBLE-STRAND BREAKS AND GENOMIC REARRANGEMENT BY GENOME-WISE INSERT CAPTURE SEQUENCING; US application 62/096,761, 24-Dec-14, ENGINEERING OF SYSTEMS, METHODS AND OPTIMIZED ENZYME AND GUIDE SCAFFOLDS FOR SEQUENCE MANIPULATION; US application 62/098,059, 30-Dec-14, 62/181,641, 18-Jun-2015, and 62/181,667, 18-Jun-2015, RNA-TARGETING SYSTEM; US application 62/096,656, 24-Dec-14 and 62/181,151, 17-Jun-2015, CRISPR HAVING OR ASSOCIATED WITH DESTABILIZATION DOMAINS; US application 62/096,697, 24-Dec-14, CRISPR HAVING OR ASSOCIATED WITH AAV; US application

62/098,158, 30-Dec-14, ENGINEERED CRISPR COMPLEX INSERTIONAL TARGETING SYSTEMS; US application 62/151,052, 22-Apr-15, CELLULAR TARGETING FOR EXTRACELLULAR EXOSOMAL REPORTING; US application 62/054,490, 24-Sep-14, DELIVERY, USE AND THERAPEUTIC APPLICATIONS OF THE CRISPR-CAS SYSTEMS AND COMPOSITIONS FOR TARGETING DISORDERS AND DISEASES USING PARTICLE DELIVERY COMPONENTS; US application 61/939,154, 12-F EB-14, SYSTEMS, METHODS AND COMPOSITIONS FOR SEQUENCE MANIPULATION WITH OPTIMIZED FUNCTIONAL CRISPR-CAS SYSTEMS; US application 62/055,484, 25-Sep-14, SYSTEMS, METHODS AND COMPOSITIONS FOR SEQUENCE MANIPULATION WITH OPTIMIZED FUNCTIONAL CRISPR-CAS SYSTEMS; US application 62/087,537, 4-Dec-14, SYSTEMS, METHODS AND COMPOSITIONS FOR SEQUENCE MANIPULATION WITH OPTIMIZED FUNCTIONAL CRISPR-CAS SYSTEMS; US application 62/054,651, 24-Sep-14, DELIVERY, USE AND THERAPEUTIC APPLICATIONS OF THE CRISPR-CAS SYSTEMS AND COMPOSITIONS FOR MODELING COMPETITION OF MULTIPLE CANCER MUTATIONS IN VIVO; US application 62/067,886, 23-Oct-14, DELIVERY, USE AND THERAPEUTIC APPLICATIONS OF THE CRISPR-CAS SYSTEMS AND COMPOSITIONS FOR MODELING COMPETITION OF MULTIPLE CANCER MUTATIONS IN VIVO; US applications 62/054,675, 24-Sep-14 and 62/181,002, 17-Jun-2015, DELIVERY, USE AND THERAPEUTIC APPLICATIONS OF THE CRISPR-CAS SYSTEMS AND COMPOSITIONS IN NEURONAL CELLS/TISSUES; US application 62/054,528, 24-Sep-14, DELIVERY, USE AND THERAPEUTIC APPLICATIONS OF THE CRISPR-CAS SYSTEMS AND COMPOSITIONS IN IMMUNE DISEASES OR DISORDERS; US application 62/055,454, 25-Sep-14, DELIVERY, USE AND THERAPEUTIC APPLICATIONS OF THE CRISPR-CAS SYSTEMS AND COMPOSITIONS FOR TARGETING DISORDERS AND DISEASES USING CELL PENETRATION PEPTIDES (CPP); US application 62/055,460, 25-Sep-14, MULTIFUNCTIONAL-CRISPR COMPLEXES AND/OR OPTIMIZED ENZYME LINKED FUNCTIONAL-CRISPR COMPLEXES; US application 62/087,475, 4-Dec-14 and 62/181,690, 18-Jun-2015, FUNCTIONAL SCREENING WITH OPTIMIZED FUNCTIONAL CRISPR-CAS SYSTEMS; US application 62/055,487, 25-Sep-14, FUNCTIONAL SCREENING WITH OPTIMIZED FUNCTIONAL CRISPR-CAS SYSTEMS; US application 62/087,546, 4-Dec-14 and 62/181,687, 18-Jun-2015, MULTIFUNCTIONAL CRISPR COMPLEXES

AND/OR OPTIMIZED ENZYME LINKED FUNCTIONAL-CRISPR COMPLEXES; and US application 62/098,285, 30-Dec-14, CRISPR MEDIATED IN VIVO MODELING AND GENETIC SCREENING OF TUMOR GROWTH AND METASTASIS.

[00280] Mention is made of US applications 62/181,659, 18-Jun-2015 and 62/207,318, 19-Aug-2015, ENGINEERING AND OPTIMIZATION OF SYSTEMS, METHODS, ENZYME AND GUIDE SCAFFOLDS OF CAS9 ORTHOLOGS AND VARIANTS FOR SEQUENCE MANIPULATION. Mention is made of US applications 62/181,663, 18-Jun-2015 and 62/245,264, 22-Oct-2015, NOVEL CRISPR ENZYMES AND SYSTEMS, US applications 62/181,675, 18-Jun-2015, 62/285,349, 22-Oct-2015, 62/296,522, 17-Feb-2016, and 62/320,231, 8-Apr-2016, NOVEL CRISPR ENZYMES AND SYSTEMS, US application 62/232,067, 24-Sep-2015, US Application 14/975,085, 18-Dec-2015, European application No. 16150428.7, US application 62/205,733, 16-Aug-2015, US application 62/201,542, 5-Aug-2015, US application 62/193,507, 16-Jul-2015, and US application 62/181,739, 18-Jun-2015, each entitled NOVEL CRISPR ENZYMES AND SYSTEMS and of US application 62/245,270, 22-Oct-2015, NOVEL CRISPR ENZYMES AND SYSTEMS. Mention is also made of US application 61/939,256, 12-Feb-2014, and WO 2015/089473 (PCT/US2014/070152), 12-Dec-2014, each entitled ENGINEERING OF SYSTEMS, METHODS AND OPTIMIZED GUIDE COMPOSITIONS WITH NEW ARCHITECTURES FOR SEQUENCE MANIPULATION. Mention is also made of PCT/US2015/045504, 15-Aug-2015, US application 62/180,699, 17-Jun-2015, and US application 62/038,358, 17-Aug-2014, each entitled GENOME EDITING USING CAS9 NICKASES.

[00281] Each of these patents, patent publications, and applications, and all documents cited therein or during their prosecution ("appln cited documents") and all documents cited or referenced in the appln cited documents, together with any instructions, descriptions, product specifications, and product sheets for any products mentioned therein or in any document therein and incorporated by reference herein, are hereby incorporated herein by reference, and may be employed in the practice of the invention. All documents (e.g., these patents, patent publications and applications and the appln cited documents) are incorporated herein by reference to the same extent as if each individual document was specifically and individually indicated to be incorporated by reference.

[00282] Type-V CRISPR-Cas Protein

[00283] The application describes methods using Type-V CRISPR-Cas proteins. This is exemplified herein with Cpf1, whereby a number of orthologs or homologs have been

identified. It will be apparent to the skilled person that further orthologs or homologs can be identified and that any of the functionalities described herein may be engineered into other orthologs, incuding chimeric enzymes comprising fragments from multiple orthologs.

[00284] Computational methods of identifying novel CRISPR-Cas loci are described in EP3009511 or US2016208243 and may comprise the following steps: detecting all contigs encoding the Cas1 protein; identifying all predicted protein coding genes within 20kB of the cas1 gene; comparing the identified genes with Cas protein-specific profiles and predicting CRISPR arrays; selecting unclassified candidate CRISPR-Cas loci containing proteins larger than 500 amino acids (>500 aa); analyzing selected candidates using methods such as PSI-BLAST and HHPred to screen for known protein domains, thereby identifying novel Class 2 CRISPR-Cas loci (see also Schmakov et al. 2015, Mol Cell. 60(3):385-97). In addition to the above mentioned steps, additional analysis of the candidates may be conducted by searching metagenomics databases for additional homologs. Additionally or alternatively, to expand the search to non-autonomous CRISPR-Cas systems, the same procedure can be performed with the CRISPR array used as the seed.

[00285] In one aspect the detecting all contigs encoding the Cas1 protein is performed by GenemarkS which a gene prediction program as further described in "GeneMarkS: a self-training method for prediction of gene starts in microbial genomes. Implications for finding sequence motifs in regulatory regions." John Besemer, Alexandre Lomsadze and Mark Borodovsky, Nucleic Acids Research (2001) 29, pp 2607-2618, herein incorporated by reference.

[00286] In one aspect the identifying all predicted protein coding genes is carried out by comparing the identified genes with Cas protein-specific profiles and annotating them according to NCBI Conserved Domain Database (CDD) which is a protein annotation resource that consists of a collection of well-annotated multiple sequence alignment models for ancient domains and full-length proteins. These are available as position-specific score matrices (PSSMs) for fast identification of conserved domains in protein sequences via RPS-BLAST. CDD content includes NCBI-curated domains, which use 3D-structure information to explicitly define domain boundaries and provide insights into sequence/structure/function relationships, as well as domain models imported from a number of external source databases (Pfam, SMART, COG, PRK, TIGRFAM). In a further aspect, CRISPR arrays were predicted using a PILER-CR program which is a public domain software for finding CRISPR repeats as described in "PILER-CR: fast and accurate

identification of CRISPR repeats", Edgar, R.C., BMC Bioinformatics, Jan 20;8:18(2007), herein incorporated by reference.

[00287] In a further aspect, the case by case analysis is performed using PSI-BLAST (Position-Specific Iterative Basic Local Alignment Search Tool). PSI-BLAST derives a position-specific scoring matrix (PSSM) or profile from the multiple sequence alignment of sequences detected above a given score threshold using protein–protein BLAST. This PSSM is used to further search the database for new matches, and is updated for subsequent iterations with these newly detected sequences. Thus, PSI-BLAST provides a means of detecting distant relationships between proteins.

[00288] In another aspect, the case by case analysis is performed using HHpred, a method for sequence database searching and structure prediction that is as easy to use as BLAST or PSI-BLAST and that is at the same time much more sensitive in finding remote homologs. In fact, HHpred's sensitivity is competitive with the most powerful servers for structure prediction currently available. HHpred is the first server that is based on the pairwise comparison of profile hidden Markov models (HMMs). Whereas most conventional sequence search methods search sequence databases such as UniProt or the NR, HHpred searches alignment databases, like Pfam or SMART. This greatly simplifies the list of hits to a number of sequence families instead of a clutter of single sequences. All major publicly available profile and alignment databases are available through HHpred. HHpred accepts a single query sequence or a multiple alignment as input. Within only a few minutes it returns the search results in an easy-to-read format similar to that of PSI-BLAST. Search options include local or global alignment and scoring secondary structure similarity. HHpred can produce pairwise query-template sequence alignments, merged query-template multiple alignments (e.g. for transitive searches), as well as 3D structural models calculated by the MODELLER software from HHpred alignments.

[00289] **Orthologs of Cpf1**

[00290] The terms "orthologue" (also referred to as "ortholog" herein) and "homologue" (also referred to as "homolog" herein) are well known in the art. By means of further guidance, a "homologue" of a protein as used herein is a protein of the same species which performs the same or a similar function as the protein it is a homologue of. Homologous proteins may but need not be structurally related, or are only partially structurally related. An "orthologue" of a protein as used herein is a protein of a different species which performs the same or a similar function as the protein it is an orthologue of. Orthologous proteins may but need not be structurally related, or are only partially structurally related. Homologs and orthologs may be

identified by homology modelling (see, e.g., Greer, Science vol. 228 (1985) 1055, and Blundell et al. Eur J Biochem vol 172 (1988), 513) or "structural BLAST" (Dey F, Cliff Zhang Q, Petrey D, Honig B. Toward a "structural BLAST": using structural relationships to infer function. Protein Sci. 2013 Apr;22(4):359-66. doi: 10.1002/pro.2225.). See also Shmakov et al. (2015) for application in the field of CRISPR-Cas loci. Homologous proteins may but need not be structurally related, or are only partially structurally related.

[00291] The Cpf1 gene is found in several diverse bacterial genomes, typically in the same locus with cas1, cas2, and cas4 genes and a CRISPR cassette (for example, FNFX1_1431-FNFX1_1428 of Francisella cf . novicida Fx1). Thus, the layout of this putative novel CRISPR-Cas system appears to be similar to that of type II-B. Furthermore, similar to Cas9, the Cpf1 protein contains a readily identifiable C-terminal region that is homologous to the transposon ORF-B and includes an active RuvC-like nuclease, an arginine-rich region, and a Zn finger (absent in Cas9). However, unlike Cas9, Cpf1 is also present in several genomes without a CRISPR-Cas context and its relatively high similarity with ORF-B suggests that it might be a transposon component. It was suggested that if this was a genuine CRISPR-Cas system and Cpf1 is a functional analog of Cas9 it would be a novel CRISPR-Cas type, namely type V (See Annotation and Classification of CRISPR-Cas Systems. Makarova KS, Koonin EV. Methods Mol Biol. 2015;1311:47-75). However, as described herein, Cpf1 is denoted to be in subtype V-A to distinguish it from C2c1p which does not have an identical domain structure and is hence denoted to be in subtype V-B.

[00292] The present invention encompasses the use of a Cpf1 effector protein, derived from a Cpf1 locus denoted as subtype V-A. Herein such effector proteins are also referred to as “Cpf1p”, e.g., a Cpf1 protein (and such effector protein or Cpf1 protein or protein derived from a Cpf1 locus is also called “CRISPR-Cas protein”).

[00293] In particular embodiments, the effector protein is a Cpf1 effector protein from an organism from a genus comprising *Streptococcus, Campylobacter, Nitratifractor, Staphylococcus, Parvibaculum, Roseburia, Neisseria, Gluconacetobacter, Azospirillum, Sphaerochaeta, Lactobacillus, Eubacterium, Corynebacter, Carnobacterium, Rhodobacter, Listeria, Paludibacter, Clostridium, Lachnospiraceae, Clostridiaridium, Leptotrichia, Francisella, Legionella, Alicyclobacillus, Methanomethyophilus, Porphyromonas, Prevotella, Bacteroidetes, Helcococcus, Leptospira, Desulfovibrio, Desulfonatronum, Opitutaceae, Tuberibacillus, Bacillus, Brevibacilus, Methylobacterium, Butyvibrio, Perigrinibacterium, Pareubacterium, Moraxella, Thiomicrospira or Acidaminococcus*. In particular embodiments, the Cpf1 effector protein is selected from an organism from a

genus selected from *Eubacterium*, *Lachnospiraceae*, *Leptotrichia*, *Francisella*, *Methanomethyophilus*, *Porphyromonas*, *Prevotella*, *Leptospira*, *Butyvibrio*, *Perigrinibacterium*, *Pareubacterium*, *Moraxella*, *Thiomicrospira or Acidaminococcus*

[00294] In further particular embodiments, the Cpf1 effector protein is from an organism selected from *S. mutans*, *S. agalactiae*, *S. equisimilis*, *S. sanguinis*, *S. pneumonia*; *C. jejuni*, *C. coli*; *N. salsuginis*, *N. tergarcus*; *S. auricularis*, *S. carnosus*; *N. meningitides*, *N. gonorrhoeae*; *L. monocytogenes*, *L. ivanovii*; *C. botulinum*, *C. difficile*, *C. tetani*, *C. sordellii*, *L inadai*, *F. tularensis 1*, *P. albensis*, *L. bacterium*, *B. proteoclasticus*, *P. bacterium*, *P. crevioricanis*, *P. disiens* and *P. macacae* .

[00295] The effector protein may comprise a chimeric effector protein comprising a first fragment from a first effector protein (e.g., a Cpf1) ortholog and a second fragment from a second effector (e.g., a Cpf1) protein ortholog, and wherein the first and second effector protein orthologs are different. At least one of the first and second effector protein (e.g., a Cpf1) orthologs may comprise an effector protein (e.g., a Cpf1) from an organism comprising *Streptococcus*, *Campylobacter*, *Nitratifractor*, *Staphylococcus*, *Parvibaculum*, *Roseburia*, *Neisseria*, *Gluconacetobacter*, *Azospirillum*, *Sphaerochaeta*, *Lactobacillus*, *Eubacterium*, *Corynebacter*, *Carnobacterium*, *Rhodobacter*, *Listeria*, *Paludibacter*, *Clostridium*, *Lachnospiraceae*, *Clostridiaridium*, *Leptotrichia*, *Francisella*, *Legionella*, *Alicyclobacillus*, *Methanomethyophilus*, *Porphyromonas*, *Prevotella*, *Bacteroidetes*, *Helcococcus*, *Letospira*, *Desulfovibrio*, *Desulfonatronum*, *Opitutaceae*, *Tuberibacillus*, *Bacillus*, *Brevibacilus*, *Methylobacterium*, *Butyvibrio*, *Perigrinibacterium*, *Pareubacterium*, *Moraxella*, *Thiomicrospira or Acidaminococcus*; e.g., a chimeric effector protein comprising a first fragment and a second fragment wherein each of the first and second fragments is selected from a Cpf1 of an organism comprising *Streptococcus*, *Campylobacter*, *Nitratifractor*, *Staphylococcus*, *Parvibaculum*, *Roseburia*, *Neisseria*, *Gluconacetobacter*, *Azospirillum*, *Sphaerochaeta*, *Lactobacillus*, *Eubacterium*, *Corynebacter*, *Carnobacterium*, *Rhodobacter*, *Listeria*, *Paludibacter*, *Clostridium*, *Lachnospiraceae*, *Clostridiaridium*, *Leptotrichia*, *Francisella*, *Legionella*, *Alicyclobacillus*, *Methanomethyophilus*, *Porphyromonas*, *Prevotella*, *Bacteroidetes*, *Helcococcus*, *Letospira*, *Desulfovibrio*, *Desulfonatronum*, *Opitutaceae*, *Tuberibacillus*, *Bacillus*, *Brevibacilus*, *Methylobacterium*, *Butyvibrio*, *Perigrinibacterium*, *Pareubacterium*, *Moraxella*, *Thiomicrospira or Acidaminococcus* wherein the first and second fragments are not from the same bacteria; for instance a chimeric effector protein comprising a first fragment and a second fragment wherein each of the first and second fragments is selected from a Cpf1 of

S. mutans, S. agalactiae, S. equisimilis, S. sanguinis, S. pneumonia; C. jejuni, C. coli; N. salsuginis, N. tergarcus; S. auricularis, S. carnosus; N. meningitides, N. gonorrhoeae; L. monocytogenes, L. ivanovii; C. botulinum, C. difficile, C. tetani, C. sordellii; Francisella tularensis 1, Prevotella albensis, Lachnospiraceae bacterium MC2017 1, Butyrivibrio proteoclasticus, Peregrinibacteria bacterium GW2011_GWA2_33_10, Parcubacteria bacterium GW2011_GWC2_44_17, Smithella sp. SCADC, Acidaminococcus sp. BV3L6, Lachnospiraceae bacterium MA2020, Candidatus Methanoplasma termitum, Eubacterium eligens, Moraxella bovoculi 237, Leptospira inadai, Lachnospiraceae bacterium ND2006, Porphyromonas crevioricanis 3, Prevotella disiens and *Porphyromonas macacae*, wherein the first and second fragments are not from the same bacteria.

[00296] In a more preferred embodiment, the Cpf1p is derived from a bacterial species selected from *Francisella tularensis 1, Prevotella albensis, Lachnospiraceae bacterium MC2017 1, Butyrivibrio proteoclasticus, Peregrinibacteria bacterium GW2011_GWA2_33_10, Parcubacteria bacterium GW2011_GWC2_44_17, Smithella sp. SCADC, Acidaminococcus sp. BV3L6, Lachnospiraceae bacterium MA2020, Candidatus Methanoplasma termitum, Eubacterium eligens, Moraxella bovoculi 237, Moraxella bovoculi AAX08_00205, Moraxella bovoculi AAX11_00205, Butyrivibrio sp. NC3005, Thiomicrospira sp. XS5, Leptospira inadai, Lachnospiraceae bacterium ND2006, Porphyromonas crevioricanis 3, Prevotella disiens* and *Porphyromonas macacae*. In certain embodiments, the Cpf1p is derived from a bacterial species selected from *Acidaminococcus sp. BV3L6, Lachnospiraceae bacterium MA2020.*In certain embodiments, the effector protein is derived from a subspecies of *Francisella tularensis 1*, including but not limited to *Francisella tularensis subsp. Novicida.* In certain preferred embodiments, the Cpf1p is derived from a bacterial species selected from *Acidaminococcus sp. BV3L6, Lachnospiraceae bacterium ND2006, Lachnospiraceae bacterium MA2020, Moraxella bovoculi AAX08_00205, Moraxella bovoculi AAX11_00205, Butyrivibrio sp. NC3005,* or *Thiomicrospira sp. XS5.*

[00297] In particular embodiments, the homologue or orthologue of Cpf1 as referred to herein has a sequence homology or identity of at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at least 95% with Cpf1. In further embodiments, the homologue or orthologue of Cpf1 as referred to herein has a sequence identity of at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at least 95% with the wild type Cpf1. Where the Cpf1 has one or more mutations (mutated), the homologue or orthologue of said Cpf1 as referred to herein has a

sequence identity of at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at least 95% with the mutated Cpf1.

[00298] In an ambodiment, the Cpf1 protein may be an ortholog of an organism of a genus which includes, but is not limited to Acidaminococcus sp, *Lachnospiraceae bacterium or Moraxella bovoculi*; in particular embodiments, the type V Cas protein may be an ortholog of an organism of a species which includes, but is not limited to *Acidaminococcus sp. BV3L6*; *Lachnospiraceae bacterium ND2006* (LbCpf1) or *Moraxella bovoculi 237*.In particular embodiments, the homologue or orthologue of Cpf1 as referred to herein has a sequence homology or identity of at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at least 95% with one or more of the Cpf1 sequences disclosed herein. In further embodiments, the homologue or orthologue of Cpf as referred to herein has a sequence identity of at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at least 95% with the wild type FnCpf1, AsCpf1 or LbCpf1.

[00299] In particular embodiments, the Cpf1 protein of the invention has a sequence homology or identity of at least 60%, more particularly at least 70, such as at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at least 95% with FnCpf1, AsCpf1 or LbCpf1. In further embodiments, the Cpf1 protein as referred to herein has a sequence identity of at least 60%, such as at least 70%, more particularly at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at least 95% with the wild type AsCpf1 or LbCpf1. In particular embodiments, the Cpf1 protein of the present invention has less than 60% sequence identity with FnCpf1. The skilled person will understand that this includes truncated forms of the Cpf1 protein whereby the sequence identity is determined over the length of the truncated form. In particular embodiments, the Cpf1 enzyme is not FnCpf1.

[00300] In some embodiments, the CRISPR effector protein is a Cpf1 protein derived from an organism from the genus of *Eubacterium*. In some embodiments, the CRISPR effector protein is a Cpf1 protein derived from an organism from the bacterial species of *Eubacterium rectale*. In some embodiments, the amino acid sequence of the Cpf1 effector protein corresponds to NCBI Reference Sequence WP_055225123.1, NCBI Reference Sequence WP_055237260.1, NCBI Reference Sequence WP_055272206.1, or GenBank ID OLA16049.1. In some embodiments, the Cpf1 effector protein has a sequence homology or sequence identity of at least 60%, more particularly at least 70%, such as at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at

least 95%, with NCBI Reference Sequence WP_055225123.1, NCBI Reference Sequence WP_055237260.1, NCBI Reference Sequence WP_055272206.1, or GenBank ID OLA16049.1. The skilled person will understand that this includes truncated forms of the Cpf1 protein whereby the sequence identity is determined over the length of the truncated form. In some embodiments, the Cpf1 effector recognizes the PAM sequence of TTTN or CTTN.

[00301] In some embodiments, the CRISPR effector protein is a Cpf1 protein derived from an organism from the genus of Eubacterium. In some embodiments, the CRISPR effector protein is a Cpf1 protein derived from an organism from the bacterial species of Eubacterium rectale. In some embodiments, the amino acid sequence of the Cpf1 effector protein corresponds to NCBI Reference Sequence WP_055225123.1, NCBI Reference Sequence WP_055237260.1, NCBI Reference Sequence WP_055272206.1, or GenBank ID OLA16049.1. In some embodiments, the Cpf1 effector protein has a sequence homology or sequence identity of at least 60%, more particularly at least 70, such as at least 80%, more preferably at least 85%, even more preferably at least 90%, such as for instance at least 95%, with NCBI Reference Sequence WP_055225123.1, NCBI Reference Sequence WP_055237260.1, NCBI Reference Sequence WP_055272206.1, or GenBank ID OLA16049.1. The skilled person will understand that this includes truncated forms of the Cpf1 protein whereby the sequence identity is determined over the length of the truncated form. In some embodiments, the Cpf1 effector recognizes the PAM sequence of TTTN or CTTN.

[00302] Codon optimized Cpf1 sequences

[00303] Where the effector protein is to be administered as a nucleic acid, the application envisages the use of codon-optimized Cpf1 sequences. An example of a codon optimized sequence, is in this instance a sequence optimized for expression in a eukaryote, e.g., humans (i.e. being optimized for expression in humans), or for another eukaryote, animal or mammal as herein discussed; see, e.g., SaCas9 human codon optimized sequence in WO 2014/093622 (PCT/US2013/074667) as an example of a codon optimized sequence (from knowledge in the art and this disclosure, codon optimizing coding nucleic acid molecule(s), especially as to effector protein (e.g., Cpf1) is within the ambit of the skilled artisan). Whilst this is preferred, it will be appreciated that other examples are possible and codon optimization for a host species other than human, or for codon optimization for specific organs is known. In some embodiments, an enzyme coding sequence encoding a DNA/RNA-targeting Cas protein is codon optimized for expression in particular cells, such

as eukaryotic cells. The eukaryotic cells may be those of or derived from a particular organism, such as a plant or a mammal, including but not limited to human, or non-human eukaryote or animal or mammal as herein discussed, e.g., mouse, rat, rabbit, dog, livestock, or non-human mammal or primate. In some embodiments, processes for modifying the germ line genetic identity of human beings and/or processes for modifying the genetic identity of animals which are likely to cause them suffering without any substantial medical benefit to man or animal, and also animals resulting from such processes, may be excluded. In general, codon optimization refers to a process of modifying a nucleic acid sequence for enhanced expression in the host cells of interest by replacing at least one codon (e.g., about or more than about 1, 2, 3, 4, 5, 10, 15, 20, 25, 50, or more codons) of the native sequence with codons that are more frequently or most frequently used in the genes of that host cell while maintaining the native amino acid sequence. Various species exhibit particular bias for certain codons of a particular amino acid. Codon bias (differences in codon usage between organisms) often correlates with the efficiency of translation of messenger RNA (mRNA), which is in turn believed to be dependent on, among other things, the properties of the codons being translated and the availability of particular transfer RNA (tRNA) molecules. The predominance of selected tRNAs in a cell is generally a reflection of the codons used most frequently in peptide synthesis. Accordingly, genes can be tailored for optimal gene expression in a given organism based on codon optimization. Codon usage tables are readily available, for example, at the "Codon Usage Database" available at www.kazusa.orjp/codon/ and these tables can be adapted in a number of ways. See Nakamura, Y., et al. "Codon usage tabulated from the international DNA sequence databases: status for the year 2000" Nucl. Acids Res. 28:292 (2000). Computer algorithms for codon optimizing a particular sequence for expression in a particular host cell are also available, such as Gene Forge (Aptagen; Jacobus, PA), are also available. In some embodiments, one or more codons (e.g., 1, 2, 3, 4, 5, 10, 15, 20, 25, 50, or more, or all codons) in a sequence encoding a DNA/RNA-targeting Cas protein corresponds to the most frequently used codon for a particular amino acid. As to codon usage in yeast, reference is made to the online Yeast Genome database available at http://www.yeastgenome.org/community/codon_usage.shtml, or Codon selection in yeast, Bennetzen and Hall, J Biol Chem. 1982 Mar 25;257(6):3026-31. As to codon usage in plants including algae, reference is made to Codon usage in higher plants, green algae, and cyanobacteria, Campbell and Gowri, Plant Physiol. 1990 Jan; 92(1): 1–11.; as well as Codon usage in plant genes, Murray et al, Nucleic Acids Res. 1989 Jan 25;17(2):477-98; or

Selection on the codon bias of chloroplast and cyanelle genes in different plant and algal lineages, Morton BR, J Mol Evol. 1998 Apr;46(4):449-59.

[00304] In certain of the following, Cpf1 amino acids are followed by *nuclear localization signals (NLS)* (italics), a glycine-serine (GS) linker (underlined), and **3x HA tag** (bold). In some embodiments, Cpf1 amino acid sequences correspond to sequences without the NLS, GS linker and 3x HA tag.

[00305] 1- Franscisella tularensis subsp. novicida U112 (FnCpf1)

[00306] MSIYQEFVNKYSLSKTLRFELIPQGKTLENIKARGLILDDEKRAKDYKKA KQIIDKYHQFFIEEILSSVCISEDLLQNYSDVYFKLKKSDDDNLQKDFKSAKDTIKKQ ISEYIKDSEKFKNLFNQNLIDAKKGQESDLILWLKQSKDNGIELFKANSDITDIDEAL EIIKSFKGWTTYFKGFHENRKNVYSSNDIPTSIIYRIVDDNLPKFLENKAKYESLKDK APEAINYEQIKKDLAEELTFDIDYKTSEVNQRVFSLDEVFEIANFNNYLNQSGITKFN TIIGGKFVNGENTKRKGINEYINLYSQQINDKTLKKYKMSVLFKQILSDTESKSFVID KLEDDSDVVTTMQSFYEQIAAFKTVEEKSIKETLSLLFDDLKAQKLDLSKIYFKNDK SLTDLSQQVFDDYSVIGTAVLEYITQQIAPKNLDNPSKKEQELIAKKTEKAKYLSLE TIKLALEEFNKHRDIDKQCRFEEILANFAAIPMIFDEIAQNKDNLAQISIKYQNQGKK DLLQASAEDDVKAIKDLLDQTNNLLHKLKIFHISQSEDKANILDKDEHFYLVFEECY FELANIVPLYNKIRNYITQKPYSDEKFKLNFENSTLANGWDKNKEPDNTAILFIKDD KYYLGVMNKKNNKIFDDKAIKENKGEGYKKIVYKLLPGANKMLPKVFFSAKSIKF YNPSEDILRIRNHSTHTKNGSPQKGYEKFEFNIEDCRKFIDFYKQSISKHPEWKDFGF RFSDTQRYNSIDEFYREVENQGYKLTFENISESYIDSVVNQGKLYLFQIYNKDFSAY SKGRPNLHTLYWKALFDERNLQDVVYKLNGEAELFYRKQSIPKKITHPAKEAIANK NKDNPKKESVFEYDLIKDKRFTEDKFFFHCPITINFKSSGANKFNDEINLLLKEKAND VHILSIDRGERHLAYYTLVDGKGNIIKQDTFNIIGNDRMKTNYHDKLAAIEKDRDSA RKDWKKINNIKEMKEGYLSQVVHEIAKLVIEYNAIVVFEDLNFGFKRGRFKVEKQV YQKLEKMLIEKLNYLVFKDNEFDKTGGVLRAYQLTAPFETFKKMGKQTGIIYYVPA GFTSKICPVTGFVNQLYPKYESVSKSQEFFSKFDKICYNLDKGYFEFSFDYKNFGDK AAKGKWTIASFGSRLINFRNSDKNHNWDTREVYPTKELEKLLKDYSIEYGHGECIK AAICGESDKKFFAKLTSVLNTILQMRNSKTGTELDYLISPVADVNGNFFDSRQAPKN MPQDADANGAYHIGLKGLMLLGRIKNNQEGKKLNLVIKNEEYFEFVQNRNN*KRPA ATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID NO:54)

[00307] 3- Lachnospiraceae bacterium MC2017 (Lb3Cpf1)

[00308] MDYGNGQFERRAPLTKTITLRLKPIGETRETIREQKLLEQDAAFRKLVET VTPIVDDCIRKIADNALCHFGTEYDFSCLGNAISKNDSKAIKKETEKVEKLLAKVLT

ENLPDGLRKVNDINSAAFIQDTLTSFVQDDADKRVLIQELKGKTVLMQRFLTTRITA
LTVWLPDRVFENFNIFIENAEKMRILLDSPLNEKIMKFDPDAEQYASLEFYGQCLSQ
KDIDSYNLIISGIYADDEVKNPGINEIVKEYNQQIRGDKDESPLPKLKKLHKQILMPV
EKAFFVRVLSNDSDARSILEKILKDTEMLPSKIIEAMKEADAGDIAVYGSRLHELSH
VIYGDHGKLSQIIYDKESKRISELMETLSPKERKESKKRLEGLEEHIRKSTYTFDELN
RYAEKNVMAAYIAAVEESCAEIMRKEKDLRTLLSKEDVKIRGNRHNTLIVKNYFN
AWTVFRNLIRILRRKSEAEIDSDFYDVLDDSVEVLSLTYKGENLCRSYITKKIGSDLK
PEIATYGSALRPNSRWWSPGEKFNVKFHTIVRRDGRLYYFILPKGAKPVELEDMDG
DIECLQMRKIPNPTIFLPKLVFKDPEAFFRDNPEADEFVFLSGMKAPVTITRETYEAY
RYKLYTVGKLRDGEVSEEEYKRALLQVLTAYKEFLENRMIYADLNFGFKDLEEYK
DSSEFIKQVETHNTFMCWAKVSSSQLDDLVKSGNGLLFEIWSERLESYYKYGNEKV
LRGYEGVLLSILKDENLVSMRTLLNSRPMLVYRPKESSKPMVVHRDGSRVVDRFD
KDGKYIPPEVHDELYRFFNNLLIKEKLGEKARKILDNKKVKVKVLESERVKWSKFY
DEQFAVTFSVKKNADCLDTTKDLNAEVMEQYSESNRLILIRNTTDILYYLVLDKNG
KVLKQRSLNIINDGARDVDWKERFRQVTKDRNEGYNEWDYSRTSNDLKEVYLNY
ALKEIAEAVIEYNAILIIEKMSNAFKDKYSFLDDVTFKGFETKLLAKLSDLHFRGIKD
GEPCSFTNPLQLCQNDSNKILQDGVIFMVPNSMTRSLDPDTGFIFAINDHNIRTKKA
KLNFLSKFDQLKVSSEGCLIMKYSGDSLPTHNTDNRVWNCCCNHPITNYDRETKKV
EFIEEPVEELSRVLEENGIETDTELNKLNERENVPGKVVDAIYSLVLNYLRGTVSGV
AGQRAVYYSPVTGKKYDISFIQAMNLNRKCDYYRIGSKERGEWTDFVAQLIN*KRPA*
*ATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID NO:55)

[00309] 4- Butyrivibrio proteoclasticus (BpCpf1)

[00310] MLLYENYTKRNQITKSLRLELRPQGKTLRNIKELNLLEQDKAIYALLERL
KPVIDEGIKDIARDTLKNCELSFEKLYEHFLSGDKKAYAKESERLKKEIVKTLIKNLP
EGIGKISEINSAKYLNGVLYDFIDKTHKDSEEKQNILSDILETKGYLALFSKFLTSRIT
TLEQSMPKRVIENFEIYAANIPKMQDALERGAVSFAIEYESICSVDYYNQILSQEDID
SYNRLISGIMDEDGAKEKGINQTISEKNIKIKSEHLEEKPFRILKQLHKQILEEREKAF
TIDHIDSDEEVVQVTKEAFEQTKEQWENIKKINGFYAKDPGDITLFIVVGPNQTHVL
SQLIYGEHDRIRLLLEEYEKNTLEVLPRRTKSEKARYDKFVNAVPKKVAKESHTFD
GLQKMTGDDRLFILYRDELARNYMRIKEAYGTFERDILKSRRGIKGNRDVQESLVS
FYDELTKFRSALRIINSGNDEKADPIFYNTFDGIFEKANRTYKAENLCRNYVTKSPA
DDARIMASCLGTPARLRTHWWNGEENFAINDVAMIRRGDEYYYFVLTPDVKPVDL
KTKDETDAQIFVQRKGAKSFLGLPKALFKCILEPYFESPEHKNDKNCVIEEYVSKPL
TIDRRAYDIFKNGTFKKTNIGIDGLTEEKFKDDCRYLIDVYKEFIAVYTRYSCFNMS

GLKRADEYNDIGEFFSDVDTRLCTMEWIPVSFERINDMVDKKEGLLFLVRSMFLYN
RPRKPYERTFIQLFSDSNMEHTSMLLNSRAMIQYRAASLPRRVTHKKGSILVALRDS
NGEHIPMHIREAIYKMKNNFDISSEDFIMAKAYLAEHDVAIKKANEDIIRNRRYTED
KFFLSLSYTKNADISARTLDYINDKVEEDTQDSRMAVIVTRNLKDLTYVAVVDEKN
NVLEEKSLNEIDGVNYRELLKERTKIKYHDKTRLWQYDVSSKGLKEAYVELAVTQI
SKLATKYNAVVVVESMSSTFKDKFSFLDEQIFKAFEARLCARMSDLSFNTIKEGEA
GSISNPIQVSNNNGNSYQDGVIYFLNNAYTRTLCPDTGFVDVFDKTRLITMQSKRQF
FAKMKDIRIDDGEMLFTFNLEEYPTKRLLDRKEWTVKIAGDGSYFDKDKGEYVYV
NDIVREQIIPALLEDKAVFDGNMAEKFLDKTAISGKSVELIYKWFANALYGIITKKD
GEKIYRSPITGTEIDVSKNTTYNFGKKFMFKQEYRGDGDFLDAFLNYMQAQDIAV*K*
*RPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID
NO:56)

[00311] 5- Peregrinibacteria bacterium GW2011_GWA_33_10 (PeCpf1)

[00312] MSNFFKNFTNLYELSKTLRFELKPVGDTLTNMKDHLEYDEKLQTFLKDQ
NIDDAYQALKPQFDEIHEEFITDSLESKKAKEIDFSEYLDLFQEKKELNDSEKKLRNK
IGETFNKAGEKWKKEKYPQYEWKKGSKIANGADILSCQDMLQFIKYKNPEDEKIK
NYIDDTLKGFFTYFGGFNQNRANYYETKKEASTAVATRIVHENLPKFCDNVIQFKHI
IKRKKDGTVEKTERKTEYLNAYQYLKNNNKITQIKDAETEKMIESTPIAEKIFDVYY
FSSCLSQKQIEEYNRIIGHYNLLINLYNQAKRSEGKHLSANEKKYKDLPKFKTLYKQ
IGCGKKKDLFYTIKCDTEEEANKSRNEGKESHSVEEIINKAQEAINKYFKSNNDCENI
NTVPDFINYILTKENYEGVYWSKAAMNTISDKYFANYHDLQDRLKEAKVFQKADK
KSEDDIKIPEAIELSGLFGVLDSLADWQTTLFKSSILSNEDKLKIITDSQTPSEALLKMI
FNDIEKNMESFLKETNDIITLKKYKGNKEGTEKIKQWFDYTLAINRMLKYFLVKEN
KIKGNSLDTNISEALKTLIYSDDAEWFKWYDALRNYLTQKPQDEAKENKLKLNFD
NPSLAGGWDVNKECSNFCVILKDKNEKKYLAIMKKGENTLFQKEWTEGRGKNLT
KKSNPLFEINNCEILSKMEYDFWADVSKMIPKCSTQLKAVVNHFKQSDNEFIFPIGY
KVTSGEKFREECKISKQDFELNNKVFNKNELSVTAMRYDLSSTQEKQYIKAFQKEY
WELLFKQEKRDTKLTNNEIFNEWINFCNKKYSELLSWERKYKDALTNWINFCKYFL
SKYPKTTLFNYSFKESENYNSLDEFYRDVDICSYKLNINTTINKSILDRLVEEGKLYL
FEIKNQDSNDGKSIGHKNNLHTIYWNAIFENFDNRPKLNGEAEIFYRKAISKDKLGI
VKGKKTKNGTEIIKNYRFSKEKFILHVPITLNFCSNNEYVNDIVNTKFYNFSNLHFLG
IDRGEKHLAYYSLVNKNGEIVDQGTLNLPFTDKDGNQRSIKKEKYFYNKQEDKWE
AKEVDCWNYNDLLDAMASNRDMARKNWQRIGTIKEAKNGYVSLVIRKIADLAVN
NERPAFIVLEDLNTGFKRSRQKIDKSVYQKFELALAKKLNFLVDKNAKRDEIGSPTK

ALQLTPPVNNYGDIENKKQAGIMLYTRANYTSQTDPATGWRKTIYLKAGPEETTY
KKDGKIKNKSVKDQIIETFTDIGFDGKDYYFEYDKGEFVDEKTGEIKPKKWRLYSG
ENGKSLDRFRGEREKDKYEWKIDKIDIVKILDDLFVNFDKNISLLKQLKEGVELTRN
NEHGTGESLRFAINLIQQIRNTGNNERDNDFILSPVRDENGKHFDSREYWDKETKGE
KISMPSSGDANGAFNIARKGIIMNAHILANSDSKDLSLFVSDEEWDLHLNNKTEWK
KQLNIFSSRKAMA*KRKKKRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYP**
YDVPDYA (SEQ ID NO:57)

[00313] 6- Parcubacteria bacterium GWC2011_GWC2_44_17 (PbCpf1)

[00314] MENIFDQFIGKYSLSKTLRFELKPVGKTEDFLKINKVFEKDQTIDDSYNQ
AKFYFDSLHQKFIDAALASDKTSELSFQNFADVLEKQNKIILDKKREMGALRKRDK
NAVGIDRLQKEINDAEDIIQKEKEKIYKDVRTLFDNEAESWKTYYQEREVDGKKITF
SKADLKQKGADFLTAAGILKVLKYEFPEEKEKEFQAKNQPSLFVEEKENPGQKRYI
FDSFDKFAGYLTKFQQTKKNLYAADGTSTAVATRIADNFIIFHQNTKVFRDKYKNN
HTDLGFDEENIFEIERYKNCLLQREIEHIKNENSYNKIIGRINKKIKEYRDQKAKDTK
LTKSDFPFFKNLDKQILGEVEKEKQLIEKTREKTEEDVLIERFKEFIENNEERFTAAK
KLMNAFCNGEFESEYEGIYLKNKAINTISRRWFVSDRDFELKLPQQKSKNKSEKNEP
KVKKFISIAEIKNAVEELDGDIFKAVFYDKKIIAQGGSKLEQFLVIWKYEFEYLFRDI
ERENGEKLLGYDSCLKIAKQLGIFPQEKEAREKATAVIKNYADAGLGIFQMMKYFS
LDDKDRKNTPGQLSTNFYAEYDGYYKDFEFIKYYNEFRNFITKKPFDEDKIKLNFEN
GALLKGWDENKEYDFMGVILKKEGRLYLGIMHKNHRKLFQSMGNAKGDNANRY
QKMIYKQIADASKDVPRLLLTSKKAMEKFKPSQEILRIKKEKTFKRESKNFSLRDLH
ALIEYYRNCIPQYSNWSFYDFQFQDTGKYQNIKEFTDDVQKYGYKISFRDIDDEYIN
QALNEGKMYLFEVVNKDIYNTKNGSKNLHTLYFEHILSAENLNDPVFKLSGMAEIF
QRQPSVNEREKITTQKNQCILDKGDRAYKYRRYTEKKIMFHMSLVLNTGKGEIKQV
QFNKIINQRISSSDNEMRVNVIGIDRGEKNLLYYSVVKQNGEIIEQASLNEINGVNYR
DKLIEREKERLKNRQSWKPVVKIKDLKKGYISHVIHKICQLIEKYSAIVVLEDLNMR
FKQIRGGIERSVYQQFEKALIDKLGYLVFKDNRDLRAPGGVLNGYQLSAPFVSFEK
MRKQTGILFYTQAEYTSKTDPITGFRKNVYISNSASLDKIKEAVKKFDAIGWDGKE
QSYFFKYNPYNLADEKYKNSTVSKEWAIFASAPRIRRQKGEDGYWKYDRVKVNEE
FEKLLKVWNFVNPKATDIKQEIIKKEKAGDLQGEKELDGRLRNFWHSFIYLFNLVL
ELRNSFSLQIKIKAGEVIAVDEGVDFIASPVKPFFTTPNPYIPSNLCWLAVENADANG
AYNIARKGVMILKKIREHAKKDPEFKKLPNLFISNAEWDEAARDWGKYAGTTALN
LDH*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID
NO:58)

[00315] 7- Smithella sp. SC_K08D17 (SsCpf1)

[00316] MQTLFENFTNQYPVSKTLRFELIPQGKTKDFIEQKGLLKKDEDRAEKYK KVKNIIDEYHKDFIEKSLNGLKLDGLEKYKTLYLKQEKDDKDKKAFDKEKENLRK QIANAFRNNEKFKTLFAKELIKNDLMSFACEEDKKNVKEFEAFTTYFTGFHQNRAN MYVADEKRTAIASRLIHENLPKFIDNIKIFEKMKKEAPELLSPFNQTLKDMKDVIKG TTLEEIFSLDYFNKTLTQSGIDIYNSVIGGRTPEEGKTKIKGLNEYINTDFNQKQTDK KKRQPKFKQLYKQILSDRQSLSFIAEAFKNDTEILEAIEKFYVNELLHFSNEGKSTNV LDAIKNAVSNLESFNLTKMYFRSGASLTDVSRKVFGEWSIINRALDNYYATTYPIKP REKSEKYEERKEKWLKQDFNVSLIQTAIDEYDNETVKGKNSGKVIADYFAKFCDD KETDLIQKVNEGYIAVKDLLNTPCPENEKLGSNKDQVKQIKAFMDSIMDIMHFVRP LSLKDTDKEKDETFYSLFTPLYDHLTQTIALYNKVRNYLTQKPYSTEKIKLNFENST LLGGWDLNKETDNTAIILRKDNLYYLGIMDKRHNRIFRNVPKADKKDFCYEKMVY KLLPGANKMLPKVFFSQSRIQEFTPSAKLLENYANETHKKGDNFNLNHCHKLIDFF KDSINKHEDWKNFDFRFSATSTYADLSGFYHEVEHQGYKISFQSVADSFIDDLVNE GKLYLFQIYNKDFSPFSKGKPNLHTLYWKMLFDENNLKDVVYKLNGEAEVFYRKK SIAEKNTTIHKANESIINKNPDNPKATSTFNYDIVKDKRYTIDKFQFHIPITMNFKAEG IFNMNQRVNQFLKANPDINIIGIDRGERHLLYYALINQKGKILKQDTLNVIANEKQK VDYHNLLDKKEGDRATARQEWGVIETIKELKEGYLSQVIHKLTDLMIENNAIIVME DLNFGFKRGRQKVEKQVYQKFEKMLIDKLNYLVDKNKKANELGGLLNAFQLANK FESFQKMGKQNGFIFYVPAWNTSKTDPATGFIDFLKPRYENLNQAKDFFEKFDSIRL NSKADYFEFAFDFKNFTEKADGGRTKWTVCTTNEDRYAWNRALNNNRGSQEKYD ITAELKSLFDGKVDYKSGKDLKQQIASQESADFFKALMKNLSITLSLRHNNGEKGD NEQDYILSPVADSKGRFFDSRKADDDMPKNADANGAYHIALKGLWCLEQISKTDD LKKVKLAISNKEWLEFVQTLKG*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVP DYAYPYDVPDYA** (SEQ ID NO:59)

[00317] 8- Acidaminococcus sp. BV3L6 (AsCpf1)

[00318] MTQFEGFTNLYQVSKTLRFELIPQGKTLKHIQEQGFIEEDKARNDHYKEL KPIIDRIYKTYADQCLQLVQLDWENLSAAIDSYRKEKTEETRNALIEEQATYRNAIH DYFIGRTDNLTDAINKRHAEIYKGLFKAELFNGKVLKQLGTVTTTEHENALLRSFD KFTTYFSGFYENRKNVFSAEDISTAIPHRIVQDNFPKFKENCHIFTRLITAVPSLREHF ENVKKAIGIFVSTSIEEVFSFPFYNQLLTQTQIDLYNQLLGGISREAGTEKIKGLNEVL NLAIQKNDETAHIIASLPHRFIPLFKQILSDRNTLSFILEEFKSDEEVIQSFCKYKTLLR NENVLETAEALFNELNSIDLTHIFISHKKLETISSALCDHWDTLRNALYERRISELTG KITKSAKEKVQRSLKHEDINLQEIISAAGKELSEAFKQKTSEILSHAHAALDQPLPTT

LKKQEEKEILKSQLDSLLGLYHLLDWFAVDESNEVDPEFSARLTGIKLEMEPSLSFY
NKARNYATKKPYSVEKFKLNFQMPTLASGWDVNKEKNNGAILFVKNGLYYLGIM
PKQKGRYKALSFEPTEKTSEGFDKMYYDYFPDAAKMIPKCSTQLKAVTAHFQTHT
TPILLSNNFIEPLEITKEIYDLNNPEKEPKKFQTAYAKKTGDQKGYREALCKWIDFTR
DFLSKYTKTTSIDLSSLRPSSQYKDLGEYYAELNPLLYHISFQRIAEKEIMDAVETGK
LYLFQIYNKDFAKGHHGKPNLHTLYWTGLFSPENLAKTSIKLNGQAELFYRPKSRM
KRMAHRLGEKMLNKKLKDQKTPIPDTLYQELYDYVNHRLSHDLSDEARALLPNVI
TKEVSHEIIKDRRFTSDKFFFHVPITLNYQAANSPSKFNQRVNAYLKEHPETPIIGIDR
GERNLIYITVIDSTGKILEQRSLNTIQQFDYQKKLDNREKERVAARQAWSVVGTIKD
LKQGYLSQVIHEIVDLMIHYQAVVVLENLNFGFKSKRTGIAEKAVYQQFEKMLIDK
LNCLVLKDYPAEKVGGVLNPYQLTDQFTSFAKMGTQSGFLFYVPAPYTSKIDPLTG
FVDPFVWKTIKNHESRKHFLEGFDFLHYDVKTGDFILHFKMNRNLSFQRGLPGFMP
AWDIVFEKNETQFDAKGTPFIAGKRIVPVIENHRFTGRYRDLYPANELIALLEEKGIV
FRDGSNILPKLLENDDSHAIDTMVALIRSVLQMRNSNAATGEDYINSPVRDLNGVC
FDSRFQNPEWPMDADANGAYHIALKGQLLLNHLKESKDLKLQNGISNQDWLAYIQ
ELRN*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID NO:60)

[00319] 9- Lachnospiraceae bacterium MA2020 (Lb2Cpf1)

[00320] MYYESLTKQYPVSKTIRNELIPIGKTLDNIRQNNILESDVKRKQNYEHVK
GILDEYHKQLINEALDNCTLPSLKIAAEIYLKNQKEVSDREDFNKTQDLLRKEVVEK
LKAHENFTKIGKKDILDLLEKLPSISEDDYNALESFRNFYTYFTSYNKVRENLYSDK
EKSSTVAYRLINENFPKFLDNVKSYRFVKTAGILADGLGEEEQDSLFIVETFNKTLT
QDGIDTYNSQVGKINSSINLYNQKNQKANGFRKIPKMKMLYKQILSDREESFIDEFQ
SDEVLIDNVESYGSVLIESLKSSKVSAFFDALRESKGKNVYVKNDLAKTAMSNIVFE
NWRTFDDLLNQEYDLANENKKKDDKYFEKRQKELKKNKSYSLEHLCNLSEDSCN
LIENYIHQISDDIENIIINNETFLRIVINEHDRSRKLAKNRKAVKAIKDFLDSIKVLERE
LKLINSSGQELEKDLIVYSAHEELLVELKQVDSLYNMTRNYLTKKPFSTEKVKLNF
NRSTLLNGWDRNKETDNLGVLLLKDGKYYLGIMNTSANKAFVNPPVAKTEKVFK
KVDYKLLPVPNQMLPKVFFAKSNIDFYNPSSEIYSNYKKGTHKKGNMFSLEDCHNL
IDFFKESISKHEDWSKFGFKFSDTASYNDISEFYREVEKQGYKLTYTDIDETYINDLIE
RNELYLFQIYNKDFSMYSKGKLNLHTLYFMMLFDQRNIDDVVYKLNGEAEVFYRP
ASISEDELIIHKAGEEIKNKNPNRARTKETSTFSYDIVKDKRYSKDKFTLHIPITMNFG
VDEVKRFNDAVNSAIRIDENVNVIGIDRGERNLLYVVVIDSKGNILEQISLNSIINKE
YDIETDYHALLDEREGGRDKARKDWNTVENIRDLKAGYLSQVVNVVAKLVLKYN

AIICLEDLNFGFKRGRQKVEKQVYQKFEKMLIDKLNYLVIDKSREQTSPKELGGAL
NALQLTSKFKSFKELGKQSGVIYYVPAYLTSKIDPTTGFANLFYMKCENVEKSKRFF
DGFDFIRFNALENVFEFGFDYRSFTQRACGINSKWTVCTNGERIIKYRNPDKNNMFD
EKVVVVTDEMKNLFEQYKIPYEDGRNVKDMIISNEEAEFYRRLYRLLQQTLQMRN
STSDGTRDYIISPVKNKREAYFNSELSDGSVPKDADANGAYNIARKGLWVLEQIRQ
KSEGEKINLAMTNAEWLEYAQTHLL*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPY**
DVPDYAYPYDVPDYA (SEQ ID NO:61)

[00321] 10- Candidatus Methanoplasma termitum (CMtCpf1)

[00322] MNNYDEFTKLYPIQKTIRFELKPQGRTMEHLETFNFFEEDRDRAEKYKIL
KEAIDEYHKKFIDEHLTNMSLDWNSLKQISEKYYKSREEKDKKVFLSEQKRMRQEI
VSEFKKDDRFKDLFSKKLFSELLKEEIYKKGNHQEIDALKSFDKFSGYFIGLHENRK
NMYSDGDEITAISNRIVNENFPKFLDNLQKYQEARKKYPEWIIKAESALVAHNIKM
DEVFSLEYFNKVLNQEGIQRYNLALGGYVTKSGEKMMGLNDALNLAHQSEKSSKG
RIHMTPLFKQILSEKESFSYIPDVFTEDSQLLPSIGGFFAQIENDKDGNIFDRALELISS
YAEYDTERIYIRQADINRVSNVIFGEWGTLGGLMREYKADSINDINLERTCKKVDK
WLDSKEFALSDVLEAIKRTGNNDAFNEYISKMRTAREKIDAARKEMKFISEKISGDE
ESIHIIKTLLDSVQQFLHFFNLFKARQDIPLDGAFYAEFDEVHSKLFAIVPLYNKVRN
YLTKNNLNTKKIKLNFKNPTLANGWDQNKVYDYASLIFLRDGNYYLGIINPKRKK
NIKFEQGSGNGPFYRKMVYKQIPGPNKNLPRVFLTSTKGKKEYKPSKEIIEGYEADK
HIRGDKFDLDFCHKLIDFFKESIEKHKDWSKFNFYFSPTESYGDISEFYLDVEKQGY
RMHFENISAETIDEYVEKGDLFLFQIYNKDFVKAATGKKDMHTIYWNAAFSPENLQ
DVVVKLNGEAELFYRDKSDIKEIVHREGEILVNRTYNGRTPVPDKIHKKLTDYHNG
RTKDLGEAKEYLDKVRYFKAHYDITKDRRYLNDKIYFHVPLTLNFKANGKKNLNK
MVIEKFLSDEKAHIIGIDRGERNLLYYSIIDRSGKIIDQQSLNVIDGFDYREKLNQREI
EMKDARQSWNAIGKIKDLKEGYLSKAVHEITKMAIQYNAIVVMEELNYGFKRGRF
KVEKQIYQKFENMLIDKMNYLVFKDAPDESPGGVLNAYQLTNPLESFAKLGKQTGI
LFYVPAAYTSKIDPTTGFVNLFNTSSKTNAQERKEFLQKFESISYSAKDGGIFAFAFD
YRKFGTSKTDHKNVWTAYTNGERMRYIKEKKRNELFDPSKEIKEALTSSGIKYDGG
QNILPDILRSNNNGLIYTMYSSFIAAIQMRVYDGKEDYIISPIKNSKGEFFRTDPKRRE
LPIDADANGAYNIALRGELTMRAIAEKFDPDSEKMAKLELKHKDWFEFMQTRGD*K*
*RPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID
NO:62)

[00323] 11- Eubacterium eligens (EeCpf1)

[00324] MNGNRSIVYREFVGVIPVAKTLRNELRPVGHTQEHIIQNGLIQEDELRQE
KSTELKNIMDDYYREYIDKSLSGVTDLDFTLLFELMNLVQSSPSKDNKKALEKEQS
KMREQICTHLQSDSNYKNIFNAKLLKEILPDFIKNYNQYDVKDKAGKLETLALFNG
FSTYFTDFFEKRKNVFTKEAVSTSIAYRIVHENSLIFLANMTSYKKISEKALDEIEVIE
KNNQDKMGDWELNQIFNPDFYNMVLIQSGIDFYNEICGVVNAHMNLYCQQTKNN
YNLFKMRKLHKQILAYTSTSFEVPKMFEDDMSVYNAVNAFIDETEKGNIIGKLKDI
VNKYDELDEKRIYISKDFYETLSCFMSGNWNLITGCVENFYDENIHAKGKSKEEKV
KKAVKEDKYKSINDVNDLVEKYIDEKERNEFKNSNAKQYIREISNIITDTETAHLEY
DDHISLIESEEKADEMKKRLDMYMNMYHWAKAFIVDEVLDRDEMFYSDIDDIYNI
LENIVPLYNRVRNYVTQKPYNSKKIKLNFQSPTLANGWSQSKEFDNNAIILIRDNKY
YLAIFNAKNKPDKKIIQGNSDKKNDNDYKKMVYNLLPGANKMLPKVFLSKKGIET
FKPSDYIISGYNAHKHIKTSENFDISFCRDLIDYFKNSIEKHAEWRKYEFKFSATDSY
SDISEFYREVEMQGYRIDWTYISEADINKLDEEGKIYLFQIYNKDFAENSTGKENLH
TMYFKNIFSEENLKDIIIKLNGQAELFYRRASVKNPVKHKKDSVLVNKTYKNQLDN
GDVVRIPIPDDIYNEIYKMYNGYIKESDLSEAAKEYLDKVEVRTAQKDIVKDYRYT
VDKYFIHTPITINYKVTARNNVNDMVVKYIAQNDDIHVIGIDRGERNLIYISVIDSHG
NIVKQKSYNILNNYDYKKKLVEKEKTREYARKNWKSIGNIKELKEGYISGVVHEIA
MLIVEYNAIIAMEDLNYGFKRGRFKVERQVYQKFESMLINKLNYFASKEKSVDEPG
GLLKGYQLTYVPDNIKNLGKQCGVIFYVPAAFTSKIDPSTGFISAFNFKSISTNASRK
QFFMQFDEIRYCAEKDMFSFGFDYNNFDTYNITMGKTQWTVYTNGERLQSEFNNA
RRTGKTKSINLTETIKLLLEDNEINYADGHDIRIDMEKMDEDKKSEFFAQLLSLYKL
TVQMRNSYTEAEEQENGISYDKIISPVINDEGEFFDSDNYKESDDKECKMPKDADA
NGAYCIALKGLYEVLKIKSEWTEDGFDRNCLKLPHAEWLDFIQNKRYE*KRPAATKK*
*AGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID NO:63)

[00325] 12- Moraxella bovoculi 237 (MbCpf1)

[00326] MLFQDFTHLYPLSKTVRFELKPIDRTLEHIHAKNFLSQDETMADMHQKV
KVILDDYHRDFIADMMGEVKLTKLAEFYDVYLKFRKNPKDDELQKQLKDLQAVL
RKEIVKPIGNGGKYKAGYDRLFGAKLFKDGKELGDLAKFVIAQEGESSPKLAHLAH
FEKFSTYFTGFHDNRKNMYSDEDKHTAIAYRLIHENLPRFIDNLQILTTIKQKHSALY
DQIINELTASGLDVSLASHLDGYHKLLTQEGITAYNTLLGGISGEAGSPKIQGINELIN
SHHNQHCHKSERIAKLRPLHKQILSDGMSVSFLPSKFADDSEMCQAVNEFYRHYAD
VFAKVQSLFDGFDDHQKDGIYVEHKNLNELSKQAFGDFALLGRVLDGYYVDVVN
PEFNERFAKAKTDNAKAKLTKEKDKFIKGVHSLASLEQAIEHYTARHDDESVQAG
KLGQYFKHGLAGVDNPIQKIHNNHSTIKGFLERERPAGERALPKIKSGKNPEMTQLR

QLKELLDNALNVAHFAKLLTTKTTLDNQDGNFYGEFGVLYDELAKIPTLYNKVRD
YLSQKPFSTEKYKLNFGNPTLLNGWDLNKEKDNFGVILQKDGCYYLALLDKAHKK
VFDNAPNTGKSIYQKMIYKYLEVRKQFPKVFFSKEAIAINYHPSKELVEIKDKGRQR
SDDERLKLYRFILECLKIHPKYDKKFEGAIGDIQLFKKDKKGREVPISEKDLFDKING
IFSSKPKLEMEDFFIGEFKRYNPSQDLVDQYNIYKKIDSNDNRKKENFYNNHPKFKK
DLVRYYYESMCKHEEWEESFEFSKKLQDIGCYVDVNELFTEIETRRLNYKISFCNIN
ADYIDELVEQGQLYLFQIYNKDFSPKAHGKPNLHTLYFKALFSEDNLADPIYKLNG
EAQIFYRKASLDMNETTIHRAGEVLENKNPDNPKKRQFVYDIIKDKRYTQDKFMLH
VPITMNFGVQGMTIKEFNKKVNQSIQQYDEVNVIGIDRGERHLLYLTVINSKGEILE
QCSLNDITTASANGTQMTTPYHKILDKREIERLNARVGWGEIETIKELKSGYLSHVV
HQISQLMLKYNAIVVLEDLNFGFKRGRFKVEKQIYQNFENALIKKLNHLVLKDKAD
DEIGSYKNALQLTNNFTDLKSIGKQTGFLFYVPAWNTSKIDPETGFVDLLKPRYENI
AQSQAFFGKFDKICYNADKDYFEFHIDYAKFTDKAKNSRQIWTICSHGDKRYVYD
KTANQNKGAAKGINVNDELKSLFARHHINEKQPNLVMDICQNNDKEFHKSLMYLL
KTLLALRYSNASSDEDFILSPVANDEGVFFNSALADDTQPQNADANGAYHIALKGL
WLLNELKNSDDLNKVKLAIDNQTWLNFAQNR*KRPAATKKAGQAKKKK*GS**YPYDVP**
DYAYPYDVPDYAYPYDVPDYA (SEQ ID NO:64)

[00327] 13- Leptospira inadai (LiCpf1)

[00328] MEDYSGFVNIYSIQKTLRFELKPVGKTLEHIEKKGFLKKDKIRAEDYKAV
KKIIDKYHRAYIEEVFDSVLHQKKKKDKTRFSTQFIKEIKEFSELYYKTEKNIPDKER
LEALSEKLRKMLVGAFKGEFSEEVAEKYKNLFSKELIRNEIEKFCETDEERKQVSNF
KSFTTYFTGFHSNRQNIYSDEKKSTAIGYRIIHQNLPKFLDNLKIIESIQRRFKDFPWS
DLKKNLKKIDKNIKLTEYFSIDGFVNVLNQKGIDAYNTILGGKSEESGEKIQGLNEYI
NLYRQKNNIDRKNLPNVKILFKQILGDRETKSFIPEAFPDDQSVLNSITEFAKYLKLD
KKKKSIIAELKKFLSSFNRYELDGIYLANDNSLASISTFLFDDWSFIKKSVSFKYDES
VGDPKKKIKSPLKYEKEKEKWLKQKYYTISFLNDAIESYSKSQDEKRVKIRLEAYFA
EFKSKDDAKKQFDLLERIEEAYAIVEPLLGAEYPRDRNLKADKKEVGKIKDFLDSIK
SLQFFLKPLLSAEIFDEKDLGFYNQLEGYYEEIDSIGHLYNKVRNYLTGKIYSKEKFK
LNFENSTLLKGWDENREVANLCVIFREDQKYYLGVMDKENNTILSDIPKVKPNELF
YEKMVYKLIPTPHMQLPRIIFSSDNLSIYNPSKSILKIREAKSFKEGKNFKLKDCHKFI
DFYKESISKNEDWSRFDFKFSKTSSYENISEFYREVERQGYNLDFKKVSKFYIDSLVE
DGKLYLFQIYNKDFSIFSKGKPNLHTIYFRSLFSKENLKDVCLKLNGEAEMFFRKKSI
NYDEKKKREGHHPELFEKLKYPILKDKRYSEDKFQFHLPISLNFKSKERLNFNLKVN
EFLKRNKDINIIGIDRGERNLLYLVMINQKGEILKQTLLDSMQSGKGRPEINYKEKL

QEKEIERDKARKSWGTVENIKELKEGYLSIVIHQISKLMVENNAIVVLEDLNIGFKR
GRQKVERQVYQKFEKMLIDKLNFLVFKENKPTEPGGVLKAYQLTDEFQSFEKLSK
QTGFLFYVPSWNTSKIDPRTGFIDFLHPAYENIEKAKQWINKFDSIRFNSKMDWFEF
TADTRKFSENLMLGKNRVWVICTTNVERYFTSKTANSSIQYNSIQITEKLKELFVDIP
FSNGQDLKPEILRKNDAVFFKSLLFYIKTTLSLRQNNGKKGEEEKDFILSPVVDSKG
RFFNSLEASDDEPKDADANGAYHIALKGLMNLLVLNETKEENLSRPKWKIKNKDW
LEFVWERNR*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA**
(SEQ ID NO:65)

[00329] 14- Lachnospiraceae bacterium ND2006 (LbCpf1)

[00330] MSKLEKFTNCYSLSKTLRFKAIPVGKTQENIDNKRLLVEDEKRAEDYKG
VKKLLDRYYLSFINDVLHSIKLKNLNNYISLFRKKTRTEKENKELENLEINLRKEIAK
AFKGNEGYKSLFKKDIIETILPEFLDDKDEIALVNSFNGFTTAFTGFFDNRENMFSEE
AKSTSIAFRCINENLTRYISNMDIFEKVDAIFDKHEVQEIKEKILNSDYDVEDFFEGEF
FNFVLTQEGIDVYNAIIGGFVTESGEKIKGLNEYINLYNQKTKQKLPKFKPLYKQVL
SDRESLSFYGEGYTSDEEVLEVFRNTLNKNSEIFSSIKKLEKLFKNFDEYSSAGIFVK
NGPAISTISKDIFGEWNVIRDKWNAEYDDIHLKKKAVVTEKYEDDRRKSFKKIGSFS
LEQLQEYADADLSVVEKLKEIIIQKVDEIYKVYGSSEKLFDADFVLEKSLKKNDAV
VAIMKDLLDSVKSFENYIKAFFGEGKETNRDESFYGDFVLAYDILLKVDHIYDAIRN
YVTQKPYSKDKFKLYFQNPQFMGGWDKDKETDYRATILRYGSKYYLAIMDKKYA
KCLQKIDKDDVNGNYEKINYKLLPGPNKMLPKVFFSKKWMAYYNPSEDIQKIYKN
GTFKKGDMFNLNDCHKLIDFFKDSISRYPKWSNAYDFNFSETEKYKDIAGFYREVE
EQGYKVSFESASKKEVDKLVEEGKLYMFQIYNKDFSDKSHGTPNLHTMYFKLLFD
ENNHGQIRLSGGAELFMRRASLKKEELVVHPANSPIANKNPDNPKKTTTLSYDVYK
DKRFSEDQYELHIPIAINKCPKNIFKINTEVRVLLKHDDNPYVIGIDRGERNLLYIVV
VDGKGNIVEQYSLNEIINNFNGIRIKTDYHSLLDKKEKERFEARQNWTSIENIKELKA
GYISQVVHKICELVEKYDAVIALEDLNSGFKNSRVKVEKQVYQKFEKMLIDKLNY
MVDKKSNPCATGGALKGYQITNKFESFKSMSTQNGFIFYIPAWLTSKIDPSTGFVNL
LKTKYTSIADSKKFISSFDRIMYVPEEDLFEFALDYKNFSRTDADYIKKWKLYSYGN
RIRIFRNPKKNNVFDWEEVCLTSAYKELFNKYGINYQQGDIRALLCEQSDKAFYSSF
MALMSLMLQMRNSITGRTDVDFLISPVKNSDGIFYDSRNYEAQENAILPKNADANG
AYNIARKVLWAIGQFKKAEDEKLDKVKIAISNKEWLEYAQTSVKH*KRPAATKKAGQ*
*AKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID NO:66)

[00331] 15- Porphyromonas crevioricanis (PcCpf1)

[00332] MDSLKDFTNLYPVSKTLRFELKPVGKTLENIEKAGILKEDEHRAESYRRV KKIIDTYHKVFIDSSLENMAKMGIENEIKAMLQSFCELYKKDHRTEGEDKALDKIRA VLRGLIVGAFTGVCGRRENTVQNEKYESLFKEKLIKEILPDFVLSTEAESLPFSVEEA TRSLKEFDSFTSYFAGFYENRKNIYSTKPQSTAIAYRLIHENLPKFIDNILVFQKIKEPI AKELEHIRADFSAGGYIKKDERLEDIFSLNYYIHVLSQAGIEKYNALIGKIVTEGDGE MKGLNEHINLYNQQRGREDRLPLFRPLYKQILSDREQLSYLPESFEKDEELLRALKE FYDHIAEDILGRTQQLMTSISEYDLSRIYVRNDSQLTDISKKMLGDWNAIYMARER AYDHEQAPKRITAKYERDRIKALKGEESISLANLNSCIAFLDNVRDCRVDTYLSTLG QKEGPHGLSNLVENVFASYHEAEQLLSFPYPEENNLIQDKDNVVLIKNLLDNISDLQ RFLKPLWGMGDEPDKDERFYGEYNYIRGALDQVIPLYNKVRNYLTRKPYSTRKVK LNFGNSQLLSGWDRNKEKDNSCVILRKGQNFYLAIMNNRHKRSFENKMLPEYKEG EPYFEKMDYKFLPDPNKMLPKVFLSKKGIEIYKPSPKLLEQYGHGTHKKGDTFSMD DLHELIDFFKHSIEAHEDWKQFGFKFSDTATYENVSSFYREVEDQGYKLSFRKVSES YVYSLIDQGKLYLFQIYNKDFSPCSKGTPNLHTLYWRMLFDERNLADVIYKLDGKA EIFFREKSLKNDHPTHPAGKPIKKKSRQKKGEESLFEYDLVKDRRYTMDKFQFHVPI TMNFKCSAGSKVNDMVNAHIREAKDMHVIGIDRGERNLLYICVIDSRGTILDQISLN TINDIDYHDLLESRDKDRQQEHRNWQTIEGIKELKQGYLSQAVHRIAELMVAYKAV VALEDLNMGFKRGRQKVESSVYQQFEKQLIDKLNYLVDKKKRPEDIGGLLRAYQF TAPFKSFKEMGKQNGFLFYIPAWNTSNIDPTTGFVNLFHVQYENVDKAKSFFQKFD SISYNPKKDWFEFAFDYKNFTKKAEGSRSMWILCTHGSRIKNFRNSQKNGQWDSEE FALTEAFKSLFVRYEIDYTADLKTAIVDEKQKDFFVDLLKLFKLTVQMRNSWKEKD LDYLISPVAGADGRFFDTREGNKSLPKDADANGAYNIALKGLWALRQIRQTSEGGK LKLAISNKEWLQFVQERSYEKD*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVP DYAYPYDVPDYA** (SEQ ID NO:67)

[00333] 16- Prevotella disiens (PdCpf1)

[00334] MENYQEFTNLFQLNKTLRFELKPIGKTCELLEEGKIFASGSFLEKDKVRA DNVSYVKKEIDKKHKIFIEETLSSFSISNDLLKQYFDCYNELKAFKKDCKSDEEEVK KTALRNKCTSIQRAMREAISQAFLKSPQKKLLAIKNLIENVFKADENVQHFSEFTSY FSGFETNRENFYSDEEKSTSIAYRLVHDNLPIFIKNIYIFEKLKEQFDAKTLSEIFENY KLYVAGSSLDEVFSLEYFNNTLTQKGIDNYNAVIGKIVKEDKQEIQGLNEHINLYNQ KHKDRRLPFFISLKKQILSDREALSWLPDMFKNDSEVIKALKGFYIEDGFENNVLTP LATLLSSLDKYNLNGIFIRNNEALSSLSQNVYRNFSIDEAIDANAELQTFNNYELIAN ALRAKIKKETKQGRKSFEKYEEYIDKKVKAIDSLSIQEINELVENYVSEFNSNSGNM PRKVEDYFSLMRKGDFGSNDLIENIKTKLSAAEKLLGTKYQETAKDIFKKDENSKLI

KELLDATKQFQHFIKPLLGTGEEADRDLVFYGDFLPLYEKFEELTLLYNKVRNRLT
QKPYSKDKIRLCFNKPKLMTGWVDSKTEKSDNGTQYGGYLFRKKNEIGEYDYFLG
ISSKAQLFRKNEAVIGDYERLDYYQPKANTIYGSAYEGENSYKEDKKRLNKVIIAYI
EQIKQTNIKKSIIESISKYPNISDDDKVTPSSLLEKIKKVSIDSYNGILSFKSFQSVNKE
VIDNLLKTISPLKNKAEFLDLINKDYQIFTEVQAVIDEICKQKTFIYFPISNVELEKEM
GDKDKPLCLFQISNKDLSFAKTFSANLRKKRGAENLHTMLFKALMEGNQDNLDLG
SGAIFYRAKSLDGNKPTHPANEAIKCRNVANKDKVSLFTYDIYKNRRYMENKFLFH
LSIVQNYKAANDSAQLNSSATEYIRKADDLHIIGIDRGERNLLYYSVIDMKGNIVEQ
DSLNIIRNNDLETDYHDLLDKREKERKANRQNWEAVEGIKDLKKGYLSQAVHQIA
QLMLKYNAIIALEDLGQMFVTRGQKIEKAVYQQFEKSLVDKLSYLVDKKRPYNEL
GGILKAYQLASSITKNNSDKQNGFLFYVPAWNTSKIDPVTGFTDLLRPKAMTIKEA
QDFFGAFDNISYNDKGYFEFETNYDKFKIRMKSAQTRWTICTFGNRIKRKKDKNY
WNYEEVELTEEFKKLFKDSNIDYENCNLKEEIQNKDNRKFFDDLIKLLQLTLQMRN
SDDKGNDYIISPVANAEGQFFDSRNGDKKLPLDADANGAYNIARKGLWNIRQIKQT
KNDKKLNLSISSTEWLDFVREKPYLK*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPY**
DVPDYAYPYDVPDYA (SEQ ID NO:68)

[00335] 17- Porphyromonas macacae (PmCpf1)

[00336] MKTQHFFEDFTSLYSLSKTIRFELKPIGKTLENIKKNGLIRRDEQRLDDYE
KLKKVIDEYHEDFIANILSSFSFSEEILQSYIQNLSESEARAKIEKTMRDTLAKAFSED
ERYKSIFKKELVKKDIPVWCPAYKSLCKKFDNFTTSLVPFHENRKNLYTSNEITASIP
YRIVHVNLPKFIQNIEALCELQKKMGADLYLEMMENLRNVWPSFVKTPDDLCNLK
TYNHLMVQSSISEYNRFVGGYSTEDGTKHQGINEWINIYRQRNKEMRLPGLVFLHK
QILAKVDSSSFISDTLENDDQVFCVLRQFRKLFWNTVSSKEDDAASLKDLFCGLSGY
DPEAIYVSDAHLATISKNIFDRWNYISDAIRRKTEVLMPRKKESVERYAEKISKQIKK
RQSYSLAELDDLLAHYSEESLPAGFSLLSYFTSLGGQKYLVSDGEVILYEEGSNIWD
EVLIAFRDLQVILDKDFTEKKLGKDEEAVSVIKKALDSALRLRKFFDLLSGTGAEIR
RDSSFYALYTDRMDKLKGLLKMYDKVRNYLTKKPYSIEKFKLHFDNPSLLSGWDK
NKELNNLSVIFRQNGYYYLGIMTPKGKNLFKTLPKLGAEEMFYEKMEYKQIAEPM
LMLPKVFFPKKTKPAFAPDQSVVDIYNKKTFKTGQKGFNKKDLYRLIDFYKEALTV
HEWKLFNFSFSPTEQYRNIGEFFDEVREQAYKVSMVNVPASYIDEAVENGKLYLFQ
IYNKDFSPYSKGIPNLHTLYWKALFSEQNQSRVYKLCGGGELFYRKASLHMQDTTV
HPKGISIHKKNLNKKGETSLFNYDLVKDKRFTEDKFFFHVPISINYKNKKITNVNQM
VRDYIAQNDDLQIIGIDRGERNLLYISRIDTRGNLLEQFSLNVIESDKGDLRTDYQKI
LGDREQERLRRRQEWKSIESIKDLKDGYMSQVVHKICNMVVEHKAIVVLENLNLSF

MKGRKKVEKSVYEKFERMLVDKLNYLVVDKKNLSNEPGGLYAAYQLTNPLFSFE
ELHRYPQSGILFFVDPWNTSLTDPSTGFVNLLGRINYTNVGDARKFFDRFNAIRYDG
KGNILFDLDLSRFDVRVETQRKLWTLTTFGSRIAKSKKSGKWMVERIENLSLCFLEL
FEQFNIGYRVEKDLKKAILSQDRKEFYVRLIYLFNLMMQIRNSDGEEDYILSPALNE
KNLQFDSRLIEAKDLPVDADANGAYNVARKGLMVVQRIKRGDHESIHRIGRAQWL
RYVQEGIVE*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA**
(SEQ ID NO:69)

[00337] 18- Thiomicrospira sp. XS5 (TsCpf1)

[00338] MTKTFDSEFFNLYSLQKTVRFELKPVGETASFVEDFKNEGLKRVVSEDE
RRAVDYQKVKEIIDDYHRDFIEESLNYFPEQVSKDALEQAFHLYQKLKAAKVEERE
KALKEWEALQKKLREKVVKCFSDSNKARFSRIDKKELIKEDLINWLVAQNREDDIP
TVETFNNFTTYFTGFHENRKNIYSKDDHATAISFRLIHENLPKFFDNVISFNKLKEGF
PELKFDKVKEDLEVDYDLKHAFEIEYFVNFVTQAGIDQYNYLLGGKTLEDGTKKQ
GMNEQINLFKQQQTRDKARQIPKLIPLFKQILSERTESQSFIPKQFESDQELFDSLQKL
HNNCQDKFTVLQQAILGLAEADLKKVFIKTSDLNALSNTIFGNYSVFSDALNLYKES
LKTKKAQEAFEKLPAHSIHDLIQYLEQFNSSLDAEKQQSTDTVLNYFIKTDELYSRFI
KSTSEAFTQVQPLFELEALSSKRRPPESEDEGAKGQEGFEQIKRIKAYLDTLMEAVH
FAKPLYLVKGRKMIEGLDKDQSFYEAFEMAYQELESLIIPIYNKARSYLSRKPFKAD
KFKINFDNNTLLSGWDANKETANASILFKKDGLYYLGIMPKGKTFLFDYFVSSEDS
EKLKQRRQKTAEEALAQDGESYFEKIRYKLLPGASKMLPKVFFSNKNIGFYNPSDDI
LRIRNTASHTKNGTPQKGHSKVEFNLNDCHKMIDFFKSSIQKHPEWGSFGFTFSDTS
DFEDMSAFYREVENQGYVISFDKIKETYIQSQVEQGNLYLFQIYNKDFSPYSKGKPN
LHTLYWKALFEEANLNNVVAKLNGEAEIFFRRHSIKASDKVVHPANQAIDNKNPHT
EKTQSTFEYDLVKDKRYTQDKFFFHVPISLNFKAQGVSKFNDKVNGFLKGNPDVNI
IGIDRGERHLLYFTVVNQKGEILVQESLNTLMSDKGHVNDYQQKLDKKEQERDAA
RKSWTTVENIKELKEGYLSHVVHKLAHLIIKYNAIVCLEDLNFGFKRGRFKVEKQV
YQKFEKALIDKLNYLVFKEKELGEVGHYLTAYQLTAPFESFKKLGKQSGILFYVPA
DYTSKIDPTTGFVNFLDLRYQSVEKAKQLLSDFNAIRFNSVQNYFEFEIDYKKLTPK
RKVGTQSKWVICTYGDVRYQNRRNQKGHWETEEVNVTEKLKALFASDSKTTTVID
YANDDNLIDVILEQDKASFFKELLWLLKLTMTLRHSKIKSEDDFILSPVKNEQGEFY
DSRKAGEVWPKDADANGAYHIALKGLWNLQQINQWEKGKTLNLAIKNQDWFSFI
QEKPYQE*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYDVPDYAYPYDVPDYA**
(SEQ ID NO:70)

[00339] 19- Moraxella bovoculi AAX08_00205 (Mb2Cpf1)

[00340] MLFQDFTHLYPLSKTVRFELKPIGRTLEHIHAKNFLSQDETMADMYQKV KVILDDYHRDFIADMMGEVKLTKLAEFYDVYLKFRKNPKDDGLQKQLKDLQAVL RKESVKPIGSGGKYKTGYDRLFGAKLFKDGKELGDLAKFVIAQEGESSPKLAHLAH FEKFSTYFTGFHDNRKNMYSDEDKHTAIAYRLIHENLPRFIDNLQILTTIKQKHSALY DQIINELTASGLDVSLASHLDGYHKLLTQEGITAYNRIIGEVNGYTNKHNQICHKSE RIAKLRPLHKQILSDGMGVSFLPSKFADDSEMCQAVNEFYRHYTDVFAKVQSLFDG FDDHQKDGIYVEHKNLNELSKQAFGDFALLGRVLDGYYVDVVNPEFNERFAKAKT DNAKAKLTKEKDKFIKGVHSLASLEQAIEHHTARHDDESVQAGKLGQYFKHGLAG VDNPIQKIHNNHSTIKGFLERERPAGERALPKIKSGKNPEMTQLRQLKELLDNALNV AHFAKLLTTKTTLDNQDGNFYGEFGVLYDELAKIPTLYNKVRDYLSQKPFSTEKYK LNFGNPTLLNGWDLNKEKDNFGVILQKDGCYYLALLDKAHKKVFDNAPNTGKNV YQKMVYKLLPGPNKMLPKVFFAKSNLDYYNPSAELLDKYAKGTHKKGDNFNLKD CHALIDFFKAGINKHPEWQHFGFKFSPTSSYRDLSDFYREVEPQGYQVKFVDINAD YIDELVEQGKLYLFQIYNKDFSPKAHGKPNLHTLYFKALFSEDNLADPIYKLNGEA QIFYRKASLDMNETTIHRAGEVLENKNPDNPKKRQFVYDIIKDKRYTQDKFMLHVP ITMNFGVQGMTIKEFNKKVNQSIQQYDEVNVIGIDRGERHLLYLTVINSKGEILEQR SLNDITTASANGTQVTTPYHKILDKREIERLNARVGWGEIETIKELKSGYLSHVVHQI NQLMLKYNAIVVLEDLNFGFKRGRFKVEKQIYQNFENALIKKLNHLVLKDKADDEI GSYKNALQLTNNFTDLKSIGKQTGFLFYVPAWNTSKIDPETGFVDLLKPRYENIAQS QAFFGKFDKICYNTDKGYFEFHIDYAKFTDKAKNSRQKWAICSHGDKRYVYDKTA NQNKGAAKGINVNDELKSLFARYHINDKQPNLVMDICQNNDKEFHKSLMCLLKTL LALRYSNASSDEDFILSPVANDEGVFFNSALADDTQPQNADANGAYHIALKGLWLL NELKNSDDLNKVKLAIDNQTWLNFAQNR*KRPAATKKAGQAKKKK*GS**YPYDVPDYA YPYDVPDYAYPYDVPDYA** (SEQ ID NO:71)

[00341] 20- Moraxella bovoculi AAX11_00205 (Mb3Cpf1)

[00342] MLFQDFTHLYPLSKTVRFELKPIGKTLEHIHAKNFLNQDETMADMYQKV KAILDDYHRDFIADMMGEVKLTKLAEFYDVYLKFRKNPKDDGLQKQLKDLQAVL RKEIVKPIGNGGKYKAGYDRLFGAKLFKDGKELGDLAKFVIAQEGESSPKLAHLAH FEKFSTYFTGFHDNRKNMYSDEDKHTAIAYRLIHENLPRFIDNLQILATIKQKHSAL YDQIINELTASGLDVSLASHLDGYHKLLTQEGITAYNTLLGGISGEAGSRKIQGINEL INSHHNQHCHKSERIAKLRPLHKQILSDGMGVSFLPSKFADDSEVCQAVNEFYRHY ADVFAKVQSLFDGFDDYQKDGIYVEYKNLNELSKQAFGDFALLGRVLDGYYVDV VNPEFNERFAKAKTDNAKAKLTKEKDKFIKGVHSLASLEQAIEHYTARHDDESVQ AGKLGQYFKHGLAGVDNPIQKIHNNHSTIKGFLERERPAGERALPKIKSDKSPEIRQ

LKELLDNALNVAHFAKLLTTKTTLHNQDGNFYGEFGALYDELAKIATLYNKVRDY
LSQKPFSTEKYKLNFGNPTLLNGWDLNKEKDNFGVILQKDGCYYLALLDKAHKKV
FDNAPNTGKSVYQKMIYKLLPGPNKMLPKVFFAKSNLDYYNPSAELLDKYAQGTH
KKGDNFNLKDCHALIDFFKAGINKHPEWQHFGFKFSPTSSYQDLSDFYREVEPQGY
QVKFVDINADYINELVEQGQLYLFQIYNKDFSPKAHGKPNLHTLYFKALFSEDNLV
NPIYKLNGEAEIFYRKASLDMNETTIHRAGEVLENKNPDNPKKRQFVYDIIKDKRYT
QDKFMLHVPITMNFGVQGMTIKEFNKKVNQSIQQYDEVNVIGIDRGERHLLYLTVI
NSKGEILEQRSLNDITTASANGTQMTTPYHKILDKREIERLNARVGWGEIETIKELKS
GYLSHVVHQISQLMLKYNAIVVLEDLNFGFKRGRFKVEKQIYQNFENALIKKLNHL
VLKDKADDEIGSYKNALQLTNNFTDLKSIGKQTGFLFYVPAWNTSKIDPETGFVDL
LKPRYENIAQSQAFFGKFDKICYNADRGYFEFHIDYAKFNDKAKNSRQIWKICSHG
DKRYVYDKTANQNKGATIGVNVNDELKSLFTRYHINDKQPNLVMDICQNNDKEFH
KSLMYLLKTLLALRYSNASSDEDFILSPVANDEGVFFNSALADDTQPQNADANGAY
HIALKGLWLLNELKNSDDLNKVKLAIDNQTWLNFAQNR*KRPAATKKAGQAKKKK*G
S**YPYDVPDYAYPYDVPDYAYPYDVPDYA** (SEQ ID NO:72)

[00343] 21- Butyrivibrio sp. NC3005 (BsCpf1)

[00344] MYYQNLTKKYPVSKTIRNELIPIGKTLENIRKNNILESDVKRKQDYEHVK
GIMDEYHKQLINEALDNYMLPSLNQAAEIYLKKHVDVEDREEFKKTQDLLRREVT
GRLKEHENYTKIGKKDILDLLEKLPSISEEDYNALESFRNFYTYFTSYNKVRENLYS
DEEKSSTVAYRLINENLPKFLDNIKSYAFVKAAGVLADCIEEEEQDALFMVETFNM
TLTQEGIDMYNYQIGKVNSAINLYNQKNHKVEEFKKIPKMKVLYKQILSDREEVFI
GEFKDDETLLSSIGAYGNVLMTYLKSEKINIFFDALRESEGKNVYVKNDLSKTTMS
NIVFGSWSAFDELLNQEYDLANENKKKDDKYFEKRQKELKKNKSYTLEQMSNLSK
EDISPIENYIERISEDIEKICIYNGEFEKIVVNEHDSSRKLSKNIKAVKVIKDYLDSIKEL
EHDIKLINGSGQELEKNLVVYVGQEEALEQLRPVDSLYNLTRNYLTKKPFSTEKVK
LNFNKSTLLNGWDKNKETDNLGILFFKDGKYYLGIMNTTANKAFVNPPAAKTENV
FKKVDYKLLPGSNKMLPKVFFAKSNIGYYNPSTELYSNYKKGTHKKGPSFSIDDCH
NLIDFFKESIKKHEDWSKFGFEFSDTADYRDISEFYREVEKQGYKLTFTDIDESYIND
LIEKNELYLFQIYNKDFSEYSKGKLNLHTLYFMMLFDQRNLDNVVYKLNGEAEVF
YRPASIAENELVIHKAGEGIKNKNPNRAKVKETSTFSYDIVKDKRYSKYKFTLHIPIT
MNFGVDEVRRFNDVINNALRTDDNVNVIGIDRGERNLLYVVVINSEGKILEQISLNS
IINKEYDIETNYHALLDEREDDRNKARKDWNTIENIKELKTGYLSQVVNVVAKLVL
KYNAIICLEDLNFGFKRGRQKVEKQVYQKFEKMLIEKLNYLVIDKSREQVSPEKMG
GALNALQLTSKFKSFAELGKQSGIIYYVPAYLTSKIDPTTGFVNLFYIKYENIEKAKQ

FFDGFDFIRFNKKDDMFEFSFDYKSFTQKACGIRSKWIVYTNGERIIKYPNPEKNNLF
DEKVINVTDEIKGLFKQYRIPYENGEDIKEIIISKAEADFYKRLFRLLHQTLQMRNST
SDGTRDYIISPVKNDRGEFFCSEFSEGTMPKDADANGAYNIARKGLWVLEQIRQKD
EGEKVNLSMTNAEWLKYAQLHLL*KRPAATKKAGQAKKKK*GS**YPYDVPDYAYPYD**
VPDYAYPYDVPDYA (SEQ ID NO:73)

[00345] Further Cpf1 orthologs include:

[00346] NCBI WP_055225123.1

[00347] MNNGTNNFQNFIGISSLQKTLRNALIPTETTQQFIVKNGIIKEDELRGENR
QILKDIMDDYYRGFISETLSSIDDIDWTSLFEKMEIQLKNGDNKDTLIKEQTEYRKAI
HKKFANDDRFKNMFSAKLISDILPEFVIHNNNYSASEKEEKTQVIKLFSRFATSFKD
YFKNRANCFSADDISSSSCHRIVNDNAEIFFSNALVYRRIVKSLSNDDINKISGDMKD
SLKEMSLEEIYSYEKYGEFITQEGISFYNDICGKVNSFMNLYCQKNKENKNLYKLQ
KLHKQILCIADTSYEVPYKFESDEEVYQSVNGFLDNISSKHIVERLRKIGDNYNGYN
LDKIYIVSKFYESVSQKTYRDWETINTALEIHYNNILPGNGKSKADKVKKAVKNDL
QKSITEINELVSNYKLCSDDNIKAETYIHEISHILNNFEAQELKYNPEIHLVESELKAS
ELKNVLDVIMNAFHWCSVFMTEELVDKDNNFYAELEEIYDEIYPVISLYNLVRNYV
TQKPYSTKKIKLNFGIPTLADGWSKSKEYSNNAIILMRDNLYYLGIFNAKNKPDKKII
EGNTSENKGDYKKMIYNLLPGPNKMIPKVFLSSKTGVETYKPSAYILEGYKQNKHI
KSSKDFDITFCHDLIDYFKNCIAIHPEWKNFGFDFSDTSTYEDISGFYREVELQGYKI
DWTYISEKDIDLLQEKGQLYLFQIYNKDFSKKSTGNDNLHTMYLKNLFSEENLKDI
VLKLNGEAEIFFRKSSIKNPIIHKKGSILVNRTYEAEEKDQFGNQIVRKNIPENIYQEL
YKYFNDKSDKELSDEAAKLKNVVGHHEAATNIVKDYRYTYDKYFLHMPITINFKA
NKTGFINDRILQYIAKEKDLHVIGIDRGERNLIYVSVIDTCGNIVEQKSFNIVNGYDY
QIKLKQQEGARQIARKEWKEIGKIKEIKEGYLSLVIHEISKMVIKYNAIIAMEDLSYG
FKKGRFKVERQVYQKFETMLINKLNYLVFKDISITENGGLLKGYQLTYIPDKLKNV
GHQCGCIFYVPAAYTSKIDPTTGFVNIFKFKDLTVDAKREFIKKFDSIRYDSEKNLFC
FTFDYNNFITQNTVMSKSSWSVYTYGVRIKRRFVNGRFSNESDTIDITKDMEKTLE
MTDINWRDGHDLRQDIIDYEIVQHIFEIFRLTVQMRNSLSELEDRDYDRLISPVLNEN
NIFYDSAKAGDALPKDADANGAYCIALKGLYEIKQITENWKEDGKFSRDKLKISNK
DWFDFIQNKRYL (SEQ ID NO:74)

[00348] NCBI WP_055237260.1

[00349] MNNGTNNFQNFIGISSLQKTLRNALIPTETTQQFIVKNGIIKEDELRGENR
QILKDIMDDYYRGFISETLSSIDDIDWTSLFEKMEIQLKNGDNKDTLIKEQAEKRKAI
YKKFADDDRFKNMFSAKLISDILPEFVIHNNNYSASEKEEKTQVIKLFSRFATSFKD

YFKNRANCFSADDISSSSCHRIVNDNAEIFFSNALVYRRIVKNLSNDDINKISGDMK
DSLKEMSLDEIYSYEKYGEFITQEGISFYNDICGKVNSFMNLYCQKNKENKNLYKL
RKLHKQILCIADTSYEVPYKFESDEEVYQSVNGFLDNISSKHIVERLRKIGDNYNGY
NLDKIYIVSRFYESVSQKTYRDWETINTALEIHYNNILPGNGKSKADKVKKAVKND
LQKSITEINELVSNYKLCPDDNIKAETYIHEISHILNNFEAQELKYNPEIHLVESELKA
SELKNVLDVIMNAFHWCSVFMTEELVDKDNNFYAELEEIYDEIYPVISLYNLVRNY
VTQKPYSTKKIKLNFGIPTLADGWSKSKEYSNNAIILMRDNLYYLGIFNAKNKPDK
KIIEGNTSENKGDYKKMIYNLLPGPNKMIPKVFLSSKTGVETYKPSAYILEGYKQNK
HLKSSKDFDITFCRDLIDYFKNCIAIHPEWKNFGFDFSDTSTYEDISGFYREVELQGY
KIDWTYISEKDIDLLQEKGQLYLFQIYNKDFSKKSTGNDNLHTMYLKNLFSEENLK
DIVLKLNGEAEIFFRKSSIKNPIIHKKGSILVNRTYEAEEKDQFGNIQIVRKTIPENIYQ
ELYKYFNDKSDKELSDEAAKLKNVVGHHEAATNIVKDYRYTYDKYFLHMPITINF
KANKTSFINDRILQYIAKENDLHVIGIDRGERNLIYVSVIDTCGNIVEQKSFNIVNGY
DYQIKLKQQEGARQIARKEWKEIGKIKEIKEGYLSLVIHEISKMVIKYNAIIAMEDLS
YGFKKGRFKVERQVYQKFETMLINKLNYLVFKDISITENGGLLKGYQLTYIPEKLK
NVGHQCGCIFYVPAAYTSKIDPTTGFANIFKFKDLTVDAKREFIKKFDSIRYDSEKNL
FCFTFDYNNFITQNTVMSKSSWSVYTYGVRIKRRFVNGRFSNESDTIDITKDMEKTL
EMTDINWRDGHDLRQDIIDYEIVQHIFEIFKLTVQMRNSLSELEDRDYDRLISPVLNE
NNIFYDSAKAGDALPKDADANGAYCIALKGLYEIKQITENWKEDGKFSRDKLKISN
KDWFDFIQNKRYL (SEQ ID NO:75)

[00350] NCBI WP_055272206.1

[00351] MNNGTNNFQNFIGISSLQKTLRNALTPTETTQQFIVKNGIIKEDELRGENR
QILKDIMDDYYRGFISETLSSIDDIDWTSLFEKMEIQLKNGDNKDTLIKEQAEKRKAI
YKKFADDDRFKNMFSAKLISDILPEFVIHNNNYSASEKEEKTQVIKLFSRFATSFKD
YFKNRANCFSADDISSSSCHRIVNDNAEIFFSNALVYRRIVKNLSNDDINKISGDMK
DSLKKMSLEKIYSYEKYGEFITQEGISFYNDICGKVNSFMNLYCQKNKENKNLYKL
RKLHKQILCIADTSYEVPYKFESDEEVYQSVNGFLDNISSKHIVERLRKIGDNYNGY
NLDKIYIVSKFYESVSQKTYRDWETINTALEIHYNNILPGNGKSKADKVKKAVKND
LQKSITEINELVSNYKLCPDDNIKAETYIHEISHILNNFEAQELKYNPEIHLVESELKA
SELKNVLDVIMNAFHWCSVFMTEELVDKDNNFYAELEEIYDEIYPVISLYNLVRNY
VTQKPYSTKKIKLNFGIPTLADGWSKSKEYSNNAIILMRDNLYYLGIFNAKNKPEKK
IIEGNTSENKGDYKKMIYNLLPGPNKMIPKVFLSSKTGVETYKPSAYILEGYKQNKH
LKSSKDFDITFCRDLIDYFKNCIAIHPEWKNFGFDFSDTSTYEDISGFYREVELQGYKI
DWTYISEKDIDLLQEKGQLYLFQIYNKDFSKKSTGNDNLHTMYLKNLFSEENLKDV

VLKLNGEAEIFFRKSSIKNPIIHKKGSILVNRTYEAEEKDQFGNIQIVRKTIPENIYQEL
YKYFNDKSDKELSDEAAKLKNAVGHHEAATNIVKDYRYTYDKYFLHMPITINFKA
NKTSFINDRILQYIAKEKDLHVIGIDRGERNLIYVSVIDTCGNIVEQKSFNIVNGYDY
QIKLKQQEGARQIARKEWKEIGKIKEIKEGYLSLVIHEISKMVIKYNAIIAMEDLSYG
FKKGRFKVERQVYQKFETMLINKLNYLVFKDISITENGGLLKGYQLTYIPEKLKNV
GHQCGCIFYVPAAYTSKIDPTTGFVNIFKFKDLTVDAKREFIKKFDSIRYDSDKNLFC
FTFDYNNFITQNTVMSKSSWSVYTYGVRIKRRFVNGRFSNESDTIDITKDMEKTLE
MTDINWRDGHDLRQDIIDYEIVQHIFEIFKLTVQMRNSLSELEDRNYDRLISPVLNEN
NIFYDSAKAGDALPKDADANGAYCIALKGLYEIKQITENWKEDGKFSRDKLKISNK
DWFDFIQNKRYL (SEQ ID NO:76)

[00352] GenBank OLA16049.1

[00353] MNNGTNNFQNFIGISSLQKTLRNALIPTETTQQFIVKNGIIKEDELRGKNR
QILKDIMDDYYRGFISETLSSIDDIDWTSLFEKMEIQLKNGDNKDTLIKEQAEKRKAI
YKKFADDDRFKNMFSAKLISDILPEFVIHNNNYSASEKKEKTQVIKLFSRFATSFKD
YFKNRANCFSADDISSSSCHRIVNDNAEIFFSNALVYRRIVKNLSNDDINKISGDMK
DSLKEMSLEEIYSYEKYGEFITQEGISFYNDICGKVNSFMNLYCQKNKENKNLYKLR
KLHKQILCIADTSYEVPYKFESDEEVYQSVNGFLDNISSKHIVERLRKIGDNYNDYN
LDKIYIVSKFYESVSQKTYRDWETINTALEIHYNNILPGNGKSKADKVKKAVKNDL
QKSITEINELVSNYKLCSDDNIKAETYIHEISHILNNFEAHELKYNPEIHLVESELKAS
ELKNVLDIIMNAFHWCSVFMTEELVDKDNNFYAELEEIYDEIYPVISLYNLVRNYVT
QKPYSTKKIKLNFGIPTLADGWSKSKEYSNNAIILMRDNLYYLGIFNAKNKPDKKIIE
GNTSENKGDYKKMIYNLLPGPNKMIPKVFLSSKTGVETYKPSAYILEGYKQNKHLK
SSKDFDITFCHDLIDYFKNCIAIHPEWKNFGFDFSDTSAYEDISGFYREVELQGYKID
WTYISEKDIDLLQEKGQLYLFQIYNKDFSKKSTGNDNLHTMYLKNLFSEENLKDIV
LKLNGEAEIFFRKSSIKNPIIHKKGSILVNRTYEAEEKDQFGNIQIVRKTIPENIYQELY
KYFNDKSDKELSDEAAKLKNVVGHHEAATNIVKDYRYTYDKYFLHMPITINFKAN
KTSFINDRILQYIAKEKDLHVIGIDRGERNLIYVSVIDTCGNIVEQKSFNIVNGYDYQI
KLKQQEGARQIARKEWKEIGKIKEIKEGYLSLVIHEISKMVIKYNAIIAMEDLSYGFK
KGRFKVERQVYQKFETMLINKLNYLVFKDISITENGGLLKGYQLTYIPDKLKNVGH
QCGCIFYVPAAYTSKIDPTTGFVNIFKFKDLTVDAKREFIKKFDSIRYDSEKNLFCFT
FDYNNFITQNTVMSKSSWSVYTYGVRIKRRFVNGRFSNESDTIDITKDMEKTLEMT
DINWRDGHDLRQDIIDYEIVQHIFEIFKLTVQMRNSLSELEDRDYDRLISPVLNENNI
FYDSAKAGYALPKDADANGAYCIALKGLYEIKQITENWKEDGKFSRDKLKISNKD
WFDFIQNKRYL (SEQ ID NO:77)

[00354] Modified Cpf1 enzymes

[00355] In particular embodiments, it is of interest to make use of an engineered Cpf1 protein as defined herein, such as Cpf1, wherein the protein complexes with a nucleic acid molecule comprising RNA to form a CRISPR complex, wherein when in the CRISPR complex, the nucleic acid molecule targets one or more target polynucleotide loci, the protein comprises at least one modification compared to unmodified Cpf1 protein, and wherein the CRISPR complex comprising the modified protein has altered activity as compared to the complex comprising the unmodified Cpf1 protein. It is to be understood that when referring herein to CRISPR "protein", the Cpf1 protein preferably is a modified CRISPR-Cas protein (e.g. having increased or decreased (or no) enzymatic activity, such as without limitation including Cpf1. The term "CRISPR protein" may be used interchangeably with "CRISPR-Cas protein", irrespective of whether the CRISPR protein has altered, such as increased or decreased (or no) enzymatic activity, compared to the wild type CRISPR protein.

[00356] Computational analysis of the primary structure of Cpf1 nucleases reveals three distinct regions. First a C-terminal RuvC like domain, which is the only functional characterized domain. Second a N-terminal alpha-helical region and thirst a mixed alpha and beta region, located between the RuvC like domain and the alpha-helical region.

[00357] Several small stretches of unstructured regions are predicted within the Cpf1 primary structure. Unstructured regions, which are exposed to the solvent and not conserved within different Cpf1 orthologs, are preferred sides for splits and insertions of small protein sequences . In addition, these sides can be used to generate chimeric proteins between Cpf1 orthologs.

[00358] Based on the above information, mutants can be generated which lead to inactivation of the enzyme or which modify the double strand nuclease to nickase activity. In alternative embodiments, this information is used to develop enzymes with reduced off-target effects (described elsewhere herein)

[00359] In certain of the above-described Cpf1 enzymes, the enzyme is modified by mutation of one or more residues (in the RuvC domain) including but not limited to positions R909, R912, R930, R947, K949, R951, R955, K965, K968, K1000, K1002, R1003, K1009, K1017, K1022, K1029, K1035, K1054, K1072, K1086, R1094, K1095, K1109, K1118, K1142, K1150, K1158, K1159, R1220, R1226, R1242, and/or R1252 with reference to amino acid position numbering of AsCpf1 (Acidaminococcus sp. BV3L6). In

certain embodiments, the Cpf1 enzymes comprising said one or more mutations have modified, more preferably increased specificity for the target.

[00360] In certain of the above-described non-naturally-occurring CRISPR-Cas proteins, the enzyme is modified by mutation of one or more residues (in the RAD50) domain including but not limited positions K324, K335, K337, R331, K369, K370, R386, R392, R393, K400, K404, K406, K408, K414, K429, K436, K438, K459, K460, K464, R670, K675, R681, K686, K689, R699, K705, R725, K729, K739, K748, and/or K752 with reference to amino acid position numbering of AsCpf1 (Acidaminococcus sp. BV3L6). In certain embodiments, the Cpf1 enzymes comprising said one or more mutations have modified, more preferably increased specificity for the target.

[00361] In certain of the Cpf1 enzymes, the enzyme is modified by mutation of one or more residues including but not limited positions R912, T923, R947, K949, R951, R955, K965, K968, K1000, R1003, K1009, K1017, K1022, K1029, K1072, K1086, F1103, R1226, and/or R1252 with reference to amino acid position numbering of AsCpf1 (Acidaminococcus sp. BV3L6). In certain embodiments, the Cpf1 enzymes comprising said one or more mutations have modified, more preferably increased specificity for the target.

[00362] In certain embodiments, the Cpf1 enzyme is modified by mutation of one or more residues including but not limited positions R833, R836, K847, K879, K881, R883, R887, K897, K900, K932, R935, K940, K948, K953, K960, K984, K1003, K1017, R1033, R1138, R1165, and/or R1252 with reference to amino acid position numbering of LbCpf1 (Lachnospiraceae bacterium ND2006). In certain embodiments, the Cpf1 enzymes comprising said one or more mutations have modified, more preferably increased specificity for the target.

[00363] In certain embodiments, the Cpf1 enzyme is modified by mutation of one or more residues including but not limited positions K15, R18, K26, Q34, R43, K48, K51, R56, R84, K85, K87, N93, R103, N104, T118, K123, K134, R176, K177, R192, K200, K226, K273, K275, T291, R301, K307, K369, S404, V409, K414, K436, K438, K468, D482, K516, R518, K524, K530, K532, K548, K559, K570, R574, K592, D596, K603, K607, K613, C647, R681, K686, H720, K739, K748, K757, T766, K780, R790, P791, K796, K809, K815, T816, K860, R862, R863, K868, K897, R909, R912, T923, R947, K949, R951, R955, K965, K968, K1000, R1003, K1009, K1017, K1022, K1029, A1053, K1072, K1086, F1103, S1209, R1226, R1252, K1273, K1282, and/or K1288 with reference to amino acid position numbering of AsCpf1 (Acidaminococcus sp. BV3L6). In certain

embodiments, the Cpf1 enzymes comprising said one or more mutations have modified, more preferably increased specificity for the target.

[00364] In certain embodiments, the enzyme is modified by mutation of one or more residues including but not limited positions K15, R18, K26, R34, R43, K48, K51, K56, K87, K88, D90, K96, K106, K107, K120, Q125, K143, R186, K187, R202, K210, K235, K296, K298, K314, K320, K326, K397, K444, K449, E454, A483, E491, K527, K541, K581, R583, K589, K595, K597, K613, K624, K635, K639, K656, K660, K667, K671, K677, K719, K725, K730, K763, K782, K791, R800, K809, K823, R833, K834, K839, K852, K858, K859, K869, K871, R872, K877, K905, R918, R921, K932, I960, K962, R964, R968, K978, K981, K1013, R1016, K1021, K1029, K1034, K1041, K1065, K1084, and/or K1098 with reference to amino acid position numbering of FnCpf1 (Francisella novicida U112). In certain embodiments, the Cpf1 enzymes comprising said one or more mutations have modified, more preferably increased specificity for the target.

[00365] In certain embodiments, the enzyme is modified by mutation of one or more residues including but not limited positions K15, R18, K26, K34, R43, K48, K51, R56, K83, K84, R86, K92, R102, K103, K116, K121, R158, E159, R174, R182, K206, K251, K253, K269, K271, K278, P342, K380, R385, K390, K415, K421, K457, K471, A506, R508, K514, K520, K522, K538, Y548, K560, K564, K580, K584, K591, K595, K601, K634, K640, R645, K679, K689, K707, T716, K725, R737, R747, R748, K753, K768, K774, K775, K785, K787, R788, Q793, K821, R833, R836, K847, K879, K881, R883, R887, K897, K900, K932, R935, K940, K948, K953, K960, K984, K1003, K1017, R1033, K1121, R1138, R1165, K1190, K1199, and/or K1208 with reference to amino acid position numbering of LbCpf1 (Lachnospiraceae bacterium ND2006). In certain embodiments, the Cpf1 enzymes comprising said one or more mutations have modified, more preferably increased specificity for the target.

[00366] In certain embodiments, the enzyme is modified by mutation of one or more residues including but not limited positions K14, R17, R25, K33, M42, Q47, K50, D55, K85, N86, K88, K94, R104, K105, K118, K123, K131, R174, K175, R190, R198, I221, K267, Q269, K285, K291, K297, K357, K403, K409, K414, K448, K460, K501, K515, K550, R552, K558, K564, K566, K582, K593, K604, K608, K623, K627, K633, K637, E643, K780, Y787, K792, K830, Q846, K858, K867, K876, K890, R900, K901, M906, K921, K927, K928, K937, K939, R940, K945, Q975, R987, R990, K1001, R1034, I1036, R1038, R1042, K1052, K1055, K1087, R1090, K1095, N1103, K1108, K1115, K1139, K1158, R1172, K1188, K1276, R1293, A1319, K1340, K1349, and/or K1356 with

reference to amino acid position numbering of MbCpf1 (Moraxella bovoculi 237). In certain embodiments, the Cpf1 enzymes comprising said one or more mutations have modified, more preferably increased specificity for the target.

[00367] In certain of the above-described Cpf1 enzymes, the enzyme is modified by mutation of one or more residues including but not limited to positions D917, E1006, E1028, D1227, D1255A, N1257, according to FnCpf1 protein or any corresponding ortholog. In an aspect the invention provides a herein-discussed composition wherein the Cpf1 enzyme is an inactivated enzyme which comprises one or more mutations selected from the group consisting of D917A, E1006A, E1028A, D1227A, D1255A, N1257A, D917A, E1006A, E1028A, D1227A, D1255A and N1257A according to FnCpf1 protein or corresponding positions in a Cpf1 ortholog. In an aspect the invention provides a herein-discussed composition, wherein the CRISPR enzyme comprises D917, or E1006 and D917, or D917 and D1255, according to FnCpf1 protein or a corresponding position in a Cpf1 ortholog.

[00368] In one embodiment, the Cpf1 protein is modified with a mutation at S1228 (e.g., S1228A) with reference to amino acid position numbering of AsCpf1. See Yamano *et al.*, *Cell* 165:949–962 (2016), which is incorporated herein by reference in its entirety.

[00369] In certain embodiments, the Cpf1 protein has been modified to recognize a non-natural PAM, such as recognizing a PAM having a sequence or comprising a sequence YCN, YCV, AYV, TYV, RYN, RCN, TGYV, NTTN, TTN, TRTN, TYTV, TYCT, TYCN, TRTN, NTTN, TACT, TYCC, TRTC, TATV, NTTV, TTV, TSTG, TVTS, TYYS, TCYS, TBYS, TCYS, TNYS, TYYS, TNTN, TSTG, TTCC, TCCC, TATC, TGTG, TCTG, TYCV, or TCTC. In particular embodiments, said mutated Cpf1 comprises one or more mutated amino acid residue at position 11, 12, 13, 14, 15, 16, 17, 34, 36, 39, 40, 43, 46, 47, 50, 54, 57, 58, 111, 126, 127, 128, 129, 130, 131, 132, 133, 134, 135, 136, 157, 158, 159, 160, 161, 162, 163, 164, 165, 166, 167, 168, 169, 170, 171, 172, 173, 174, 175, 176, 177, 178, 532, 533, 534, 535, 536, 537, 538, 539, 540, 541, 542, 543, 544, 545, 546, 547, 548, 549, 550, 551, 552, 553, 554, 555, 556, 565, 566, 567, 568, 569, 570, 571, 572, 573, 574, 575, 592, 593, 594, 595, 596, 597, 598, 599, 600, 601, 602, 603, 604, 605, 606, 607, 608, 609, 610, 611, 612, 613, 614, 615, 616, 617, 618, 619, 620, 626, 627, 628, 629, 630, 631, 632, 633, 634, 635, 636, 637, 638, 642, 643, 644, 645, 646, 647, 648, 649, 651, 652, 653, 654, 655, 656, 676, 679, 680, 682, 683, 684, 685, 686, 687, 688, 689, 690, 691, 692, 693, 707, 711, 714, 715, 716, 717, 718, 719, 720, 721, 722, 739, 765, 768, 769, 773, 777, 778,

779, 780, 781, 782, 783, 784, 785, 786, 871, 872, 873, 874, 875, 876, 877, 878, 879, 880, 881, 882, 883, 884, or 1048 of AsCpf1 or a position corresponding thereto in a Cpf1 ortholog; preferably, one or more mutated amino acid residue at position 130, 131, 132, 133, 134, 135, 136, 162, 163, 164, 165, 166, 167, 168, 169, 170, 171, 172, 173, 174, 175, 176, 177, 536, 537, 538, 539, 540, 541, 542, 543, 544, 545, 546, 547, 548, 549, 550, 551, 552, 570, 571, 572, 573, 595, 596, 597, 598, 599, 600, 601, 602, 603, 604, 605, 606, 607, 608, 609, 610, 611, 612, 613, 614, 615, 630, 631, 632, 646, 647, 648, 649, 650, 651, 652, 653, 683, 684, 685, 686, 687, 688, 689, or 690;

[00370] In certain embodiments, the Cpf1 protein is modified to have increased activity, i.e. wider PAM specificity. In particular embodiments, the Cpf1 protein is modified by mutation of one or more residues including but not limited positions 539, 542, 547, 548, 550, 551, 552, 167, 604, and/or 607 of AsCpf1, or the corresponding position of an AsCpf1 orthologue, homologue, or variant, preferably mutated amino acid residues at positions 542 or 542 and 607, wherein said mutations preferably are 542R and 607R, such as S542R and K607R; or preferably mutated amino acid residues at positions 542 and 548 (and optionally 552), wherein said mutations preferably are 542R and 548V (and optionally 552R), such as S542R and K548V (and optionally N552R); or at position 532, 538, 542, and/or 595 of LbCpf1, or the corresponding position of an AsCpf1 orthologue, homologue, or variant, preferably mutated amino acid residues at positions 532 or 532 and 595, wherein said mutations preferably are 532R and 595R, such as G532R and K595R; or preferably mutated amino acid residues at positions 532 and 538 (and optionally 542), wherein said mutations preferably are 532R and 538V (and optionally 542R), such as G532R and K538V (and optionally Y542R), most preferably wherein said mutations are S542R and K607R, S542R and K548V, or S542R, K548V and N552R of AsCpf1.

[00371] Deactivated / inactivated Cpf1 protein

[00372] Where the Cpf1 protein has nuclease activity, the Cpf1 protein may be modified to have diminished nuclease activity e.g., nuclease inactivation of at least 70%, at least 80%, at least 90%, at least 95%, at least 97%, or 100% as compared with the wild type enzyme; or to put in another way, a Cpf1 enzyme having advantageously about 0% of the nuclease activity of the non-mutated or wild type Cpf1 enzyme or CRISPR-Cas protein, or no more than about 3% or about 5% or about 10% of the nuclease activity of the non-mutated or wild type Cpf1 enzyme, e.g. of the non-mutated or wild type *Francisella novicida U112* (FnCpf1), *Acidaminococcus sp. BV3L6* (AsCpf1), *Lachnospiraceae bacterium ND2006* (LbCpf1) or *Moraxella bovoculi 237* (MbCpf1 Cpf1 enzyme or

CRISPR-Cas protein. This is possible by introducing mutations into the nuclease domains of the Cpf1 and orthologs thereof.

[00373] In preferred embodiments of the present invention at least one Cpf1 protein is used which is a Cpf1 nickase. More particularly, a Cpf1 nickase is used which does not cleave the target strand but is capable of cleaving only the strand which is complementary to the target strand, i.e. the non-target DNA strand also referred to herein as the strand which is not complementary to the guide sequence. More particularly the Cpf1 nickase is a Cpf1 protein which comprises a mutation in the arginine at position 1226A in the Nuc domain of Cpf1 from *Acidaminococcus sp.*, or a corresponding position in a Cpf1 ortholog. In further particular embodiments, the enzyme comprises an arginine-to-alanine substitution or an R1226A mutation. It will be understood by the skilled person that where the enzyme is not AsCpf1, a mutation may be made at a residue in a corresponding position. In particular embodiments, the Cpf1 is FnCpf1 and the mutation is at the arginine at position R1218. In particular embodiments, the Cpf1 is LbCpf1 and the mutation is at the arginine at position R1138. In particular embodiments, the Cpf1 is MbCpf1 and the mutation is at the arginine at position R1293.

[00374] In certain embodiments, use is made additionally or alternatively of a CRISPR-Cas protein which is is engineered and can comprise one or more mutations that reduce or eliminate a nuclease activity. The amino acid positions in the FnCpf1p RuvC domain include but are not limited to D917A, E1006A, E1028A, D1227A, D1255A, N1257A, D917A, E1006A, E1028A, D1227A, D1255A and N1257A. Applicants have also identified a putative second nuclease domain which is most similar to PD-(D/E)XK nuclease superfamily and HincII endonuclease like. The point mutations to be generated in this putative nuclease domain to substantially reduce nuclease activity include but are not limited to N580A, N584A, T587A, W609A, D610A, K613A, E614A, D616A, K624A, D625A, K627A and Y629A. In a preferred embodiment, the mutation in the FnCpf1p RuvC domain is D917A or E1006A, wherein the D917A or E1006A mutation completely inactivates the DNA cleavage activity of the FnCpf1 effector protein. In another embodiment, the mutation in the FnCpf1p RuvC domain is D1255A, wherein the mutated FnCpf1 effector protein has significantly reduced nucleolytic activity.

[00375] More particularly, the inactivated Cpf1 enzymes include enzymes mutated in amino acid positions As908, As993, As1263 of AsCpf1 or corresponding positions in Cpf1 orthologs. Additionally, the inactivated Cpf1 enzymes include enzymes mutated in amino acid position Lb832, 925, 947 or 1180 of LbCpf1 or corresponding positions in Cpf1

orthologs. More particularly, the inactivated Cpf1 enzymes include enzymes comprising one or more of mutations AsD908A, AsE993A, AsD1263A of AsCpf1 or corresponding mutations in Cpf1 orthologs. Additionally, the inactivated Cpf1 enzymes include enzymes comprising one or more of mutations LbD832A, E925A, D947A or D1180A of LbCpf1 or corresponding mutations in Cpf1 orthologs.

[00376] Mutations can also be made at neighboring residues, e.g., at amino acids near those indicated above that participate in the nuclease acrivity. In some embodiments, only the RuvC domain is inactivated, and in other embodiments, another putative nuclease domain is inactivated, wherein the effector protein complex functions as a nickase and cleaves only one DNA strand. In a preferred embodiment, the other putative nuclease domain is a HincII-like endonuclease domain.

[00377] The inactivated Cpf1 or Cpf1 nickase may have associated (e.g., via fusion protein) one or more functional domains, including for example, an adenosine deaminase or catalytic domain thereof. In some cases it is advantageous that additionally at least one heterologous NLS is provided. In some instances, it is advantageous to position the NLS at the N terminus. In general, the positioning of the one or more functional domain on the inactivated Cpf1 or Cpf1 nickase is one which allows for correct spatial orientation for the functional domain to affect the target with the attributed functional effect. For example, when the functional domain is an adenosine deaminase catalytic domain thereof, the adenosine deaminase catalytic domain is placed in a spatial orientation which allows it to contact and deaminate a target adenine. This may include positions other than the N- / C-terminus of Cpf1. In some embodiments, the adenosine deaminase protein or catalytic domain thereof is inserted into an internal loop of Cpf1.

[00378] Determination of PAM

[00379] Determination of PAM can be ensured as follows. This experiment closely parallels similar work in *E. coli* for the heterologous expression of StCas9 (Sapranauskas, R. et al. Nucleic Acids Res 39, 9275–9282 (2011)). Applicants introduce a plasmid containing both a PAM and a resistance gene into the heterologous *E. coli*, and then plate on the corresponding antibiotic. If there is DNA cleavage of the plasmid, Applicants observe no viable colonies.

[00380] In further detail, the assay is as follows for a DNA target. Two *E.coli* strains are used in this assay. One carries a plasmid that encodes the endogenous effector protein locus from the bacterial strain. The other strain carries an empty plasmid (e.g.pACYC184, control strain). All possible 7 or 8 bp PAM sequences are presented on an antibiotic resistance

plasmid (pUC19 with ampicillin resistance gene). The PAM is located next to the sequence of proto-spacer 1 (the DNA target to the first spacer in the endogenous effector protein locus). Two PAM libraries were cloned. One has a 8 random bp 5' of the proto-spacer (e.g. total of 65536 different PAM sequences = complexity). The other library has 7 random bp 3' of the proto-spacer (e.g. total complexity is 16384 different PAMs). Both libraries were cloned to have in average 500 plasmids per possible PAM. Test strain and control strain were transformed with 5'PAM and 3'PAM library in separate transformations and transformed cells were plated separately on ampicillin plates. Recognition and subsequent cutting/interference with the plasmid renders a cell vulnerable to ampicillin and prevents growth. Approximately 12h after transformation, all colonies formed by the test and control strains where harvested and plasmid DNA was isolated. Plasmid DNA was used as template for PCR amplification and subsequent deep sequencing. Representation of all PAMs in the untransfomed libraries showed the expected representation of PAMs in transformed cells. Representation of all PAMs found in control strains showed the actual representation. Representation of all PAMs in test strain showed which PAMs are not recognized by the enzyme and comparison to the control strain allows extracting the sequence of the depleted PAM.

[00381] The following PAMs have been identified for certain wild-type Cpf1 orthologues: the Acidaminococcus sp. BV3L6 Cpf1 (AsCpf1), Lachnospiraceae bacterium ND2006 Cpf1 (LbCpf1) and Prevotella albensis (PaCpf1) can cleave target sites preceded by a TTTV PAM, where V is A/C or G, FnCpf1p, can cleave sites preceded by TTN, where N is A/C/G or T. The Moraxella bovoculi AAX08_00205, Moraxella bovoculi AAX11_00205, Butyrivibrio sp. NC3005, Thiomicrospira sp. XS5, or Lachnospiraceae bacterium MA2020 PAM is 5' TTN, where N is A/C/G or T. The natural PAM sequence is TTTV or BTTV, wherein B is T/C or G and V is A/C or G and the effector protein is Moraxella lacunata Cpf1.

[00382] **Delivery**

[00383] In some embodiments, the components of the AD-functionalized CRISPR-Cas system may be delivered in various form, such as combinations of DNA/RNA or RNA/RNA or protein RNA. For example, the Cpf1 protein may be delivered as a DNA-coding polynucleotide or an RNA-coding polynucleotide or as a protein. The guide may be delivered may be delivered as a DNA-coding polynucleotide or an RNA. All possible combinations are envisioned, including mixed forms of delivery.

[00384] In some aspects, the invention provides methods comprising delivering one or more polynucleotides, such as or one or more vectors as described herein, one or more transcripts thereof, and/or one or proteins transcribed therefrom, to a host cell.

[00385] Vectors

[00386] In general, the term "vector" refers to a nucleic acid molecule capable of transporting another nucleic acid to which it has been linked. It is a replicon, such as a plasmid, phage, or cosmid, into which another DNA segment may be inserted so as to bring about the replication of the inserted segment. Generally, a vector is capable of replication when associated with the proper control elementsVectors include, but are not limited to, nucleic acid molecules that are single-stranded, double-stranded, or partially double-stranded; nucleic acid molecules that comprise one or more free ends, no free ends (e.g., circular); nucleic acid molecules that comprise DNA, RNA, or both; and other varieties of polynucleotides known in the art. One type of vector is a "plasmid," which refers to a circular double stranded DNA loop into which additional DNA segments can be inserted, such as by standard molecular cloning techniques. Another type of vector is a viral vector, wherein virally-derived DNA or RNA sequences are present in the vector for packaging into a virus (e.g., retroviruses, replication defective retroviruses, adenoviruses, replication defective adenoviruses, and adeno-associated viruses). Viral vectors also include polynucleotides carried by a virus for transfection into a host cell. Certain vectors are capable of autonomous replication in a host cell into which they are introduced (e.g., bacterial vectors having a bacterial origin of replication and episomal mammalian vectors). Other vectors (e.g., non-episomal mammalian vectors) are integrated into the genome of a host cell upon introduction into the host cell, and thereby are replicated along with the host genome. Moreover, certain vectors are capable of directing the expression of genes to which they are operatively-linked. Such vectors are referred to herein as "expression vectors." Vectors for and that result in expression in a eukaryotic cell can be referred to herein as "eukaryotic expression vectors." Common expression vectors of utility in recombinant DNA techniques are often in the form of plasmids.

[00387] Recombinant expression vectors can comprise a nucleic acid of the invention in a form suitable for expression of the nucleic acid in a host cell, which means that the recombinant expression vectors include one or more regulatory elements, which may be selected on the basis of the host cells to be used for expression, that is operatively-linked to the nucleic acid sequence to be expressed. Within a recombinant expression vector, "operably linked" is intended to mean that the nucleotide sequence of interest is linked to

the regulatory element(s) in a manner that allows for expression of the nucleotide sequence (e.g., in an in vitro transcription/translation system or in a host cell when the vector is introduced into the host cell). Advantageous vectors include lentiviruses and adeno-associated viruses, and types of such vectors can also be selected for targeting particular types of cells.

[00388] With regards to recombination and cloning methods, mention is made of U.S. patent application 10/815,730, published September 2, 2004 as US 2004-0171156 A1, the contents of which are herein incorporated by reference in their entirety.

[00389] The term "regulatory element" is intended to include promoters, enhancers, internal ribosomal entry sites (IRES), and other expression control elements (e.g., transcription termination signals, such as polyadenylation signals and poly-U sequences). Such regulatory elements are described, for example, in Goeddel, GENE EXPRESSION TECHNOLOGY: METHODS IN ENZYMOLOGY 185, Academic Press, San Diego, Calif. (1990). Regulatory elements include those that direct constitutive expression of a nucleotide sequence in many types of host cell and those that direct expression of the nucleotide sequence only in certain host cells (e.g., tissue-specific regulatory sequences). A tissue-specific promoter may direct expression primarily in a desired tissue of interest, such as muscle, neuron, bone, skin, blood, specific organs (e.g., liver, pancreas), or particular cell types (e.g., lymphocytes). Regulatory elements may also direct expression in a temporal-dependent manner, such as in a cell-cycle dependent or developmental stage-dependent manner, which may or may not also be tissue or cell-type specific. In some embodiments, a vector comprises one or more pol III promoter (e.g., 1, 2, 3, 4, 5, or more pol III promoters), one or more pol II promoters (e.g., 1, 2, 3, 4, 5, or more pol II promoters), one or more pol I promoters (e.g., 1, 2, 3, 4, 5, or more pol I promoters), or combinations thereof. Examples of pol III promoters include, but are not limited to, U6 and H1 promoters. Examples of pol II promoters include, but are not limited to, the retroviral Rous sarcoma virus (RSV) LTR promoter (optionally with the RSV enhancer), the cytomegalovirus (CMV) promoter (optionally with the CMV enhancer) [see, e.g., Boshart et al, Cell, 41:521-530 (1985)], the SV40 promoter, the dihydrofolate reductase promoter, the β-actin promoter, the phosphoglycerol kinase (PGK) promoter, and the EF1α promoter. Also encompassed by the term "regulatory element" are enhancer elements, such as WPRE; CMV enhancers; the R-U5' segment in LTR of HTLV-I (Mol. Cell. Biol., Vol. 8(1), p. 466-472, 1988); SV40 enhancer; and the intron sequence between exons 2 and 3 of rabbit β-globin (Proc. Natl. Acad. Sci. USA., Vol. 78(3), p. 1527-31, 1981). It will be appreciated by those skilled in

the art that the design of the expression vector can depend on such factors as the choice of the host cell to be transformed, the level of expression desired, etc. A vector can be introduced into host cells to thereby produce transcripts, proteins, or peptides, including fusion proteins or peptides, encoded by nucleic acids as described herein (e.g., clustered regularly interspersed short palindromic repeats (CRISPR) transcripts, proteins, enzymes, mutant forms thereof, fusion proteins thereof, etc.). With regards to regulatory sequences, mention is made of U.S. patent application 10/491,026, the contents of which are incorporated by reference herein in their entirety. With regards to promoters, mention is made of PCT publication WO 2011/028929 and U.S. application 12/511,940, the contents of which are incorporated by reference herein in their entirety.

[00390] Advantageous vectors include lentiviruses and adeno-associated viruses, and types of such vectors can also be selected for targeting particular types of cells.

[00391] In particular embodiments, use is made of bicistronic vectors for the guide RNA and (optionally modified or mutated) the CRISPR-Cas protein fused to adenosine deaminase. Bicistronic expression vectors for guide RNA and (optionally modified or mutated) CRISPR-Cas protein fused to adenosine deaminase are preferred. In general and particularly in this embodiment, (optionally modified or mutated) CRISPR-Cas protein fused to adenosine deaminase is preferably driven by the CBh promoter. The RNA may preferably be driven by a Pol III promoter, such as a U6 promoter. Ideally the two are combined.

[00392] Vectors can be designed for expression of CRISPR transcripts (e.g. nucleic acid transcripts, proteins, or enzymes) in prokaryotic or eukaryotic cells. For example, CRISPR transcripts can be expressed in bacterial cells such as Escherichia coli, insect cells (using baculovirus expression vectors), yeast cells, or mammalian cells. Suitable host cells are discussed further in Goeddel, GENE EXPRESSION TECHNOLOGY: METHODS IN ENZYMOLOGY 185, Academic Press, San Diego, Calif. (1990). Alternatively, the recombinant expression vector can be transcribed and translated in vitro, for example using T7 promoter regulatory sequences and T7 polymerase.

[00393] Vectors may be introduced and propagated in a prokaryote or prokaryotic cell. In some embodiments, a prokaryote is used to amplify copies of a vector to be introduced into a eukaryotic cell or as an intermediate vector in the production of a vector to be introduced into a eukaryotic cell (e.g. amplifying a plasmid as part of a viral vector packaging system). In some embodiments, a prokaryote is used to amplify copies of a vector and express one or more nucleic acids, such as to provide a source of one or more

proteins for delivery to a host cell or host organism. Expression of proteins in prokaryotes is most often carried out in Escherichia coli with vectors containing constitutive or inducible promoters directing the expression of either fusion or non-fusion proteins. Fusion vectors add a number of amino acids to a protein encoded therein, such as to the amino terminus of the recombinant protein. Such fusion vectors may serve one or more purposes, such as: (i) to increase expression of recombinant protein; (ii) to increase the solubility of the recombinant protein; and (iii) to aid in the purification of the recombinant protein by acting as a ligand in affinity purification. Often, in fusion expression vectors, a proteolytic cleavage site is introduced at the junction of the fusion moiety and the recombinant protein to enable separation of the recombinant protein from the fusion moiety subsequent to purification of the fusion protein. Such enzymes, and their cognate recognition sequences, include Factor Xa, thrombin and enterokinase. Example fusion expression vectors include pGEX (Pharmacia Biotech Inc; Smith and Johnson, 1988. *Gene* 67: 31-40), pMAL (New England Biolabs, Beverly, Mass.) and pRIT5 (Pharmacia, Piscataway, N.J.) that fuse glutathione S-transferase (GST), maltose E binding protein, or protein A, respectively, to the target recombinant protein. Examples of suitable inducible non-fusion E. coli expression vectors include pTrc (Amrann et al., (1988) Gene 69:301-315) and pET 11d (Studier et al., GENE EXPRESSION TECHNOLOGY: METHODS IN ENZYMOLOGY 185, Academic Press, San Diego, Calif. (1990) 60-89). In some embodiments, a vector is a yeast expression vector. Examples of vectors for expression in yeast Saccharomyces cerivisae include pYepSec1 (Baldari, et al., 1987. EMBO J. 6: 229-234), pMFa (Kuijan and Herskowitz, 1982. Cell 30: 933-943), pJRY88 (Schultz et al., 1987. Gene 54: 113-123), pYES2 (Invitrogen Corporation, San Diego, Calif.), and picZ (InVitrogen Corp, San Diego, Calif.). In some embodiments, a vector drives protein expression in insect cells using baculovirus expression vectors. Baculovirus vectors available for expression of proteins in cultured insect cells (e.g., SF9 cells) include the pAc series (Smith, et al., 1983. Mol. Cell. Biol. 3: 2156-2165) and the pVL series (Lucklow and Summers, 1989. Virology 170: 31-39).

[00394] In some embodiments, a vector is capable of driving expression of one or more sequences in mammalian cells using a mammalian expression vector. Examples of mammalian expression vectors include pCDM8 (Seed, 1987. Nature 329: 840) and pMT2PC (Kaufman, et al., 1987. EMBO J. 6: 187-195). When used in mammalian cells, the expression vector's control functions are typically provided by one or more regulatory elements. For example, commonly used promoters are derived from polyoma, adenovirus 2, cytomegalovirus, simian virus 40, and others disclosed herein and known in the art. For

other suitable expression systems for both prokaryotic and eukaryotic cells see, e.g., Chapters 16 and 17 of Sambrook, et al., MOLECULAR CLONING: A LABORATORY MANUAL. 2nd ed., Cold Spring Harbor Laboratory, Cold Spring Harbor Laboratory Press, Cold Spring Harbor, N.Y., 1989.

[00395] In some embodiments, the recombinant mammalian expression vector is capable of directing expression of the nucleic acid preferentially in a particular cell type (e.g., tissue-specific regulatory elements are used to express the nucleic acid). Tissue-specific regulatory elements are known in the art. Non-limiting examples of suitable tissue-specific promoters include the albumin promoter (liver-specific; Pinkert, et al., 1987. *Genes Dev.* 1: 268-277), lymphoid-specific promoters (Calame and Eaton, 1988. *Adv. Immunol.* 43: 235-275), in particular promoters of T cell receptors (Winoto and Baltimore, 1989. *EMBO J.* 8: 729-733) and immunoglobulins (Baneiji, et al., 1983. *Cell* 33: 729-740; Queen and Baltimore, 1983. *Cell* 33: 741-748), neuron-specific promoters (e.g., the neurofilament promoter; Byrne and Ruddle, 1989. *Proc. Natl. Acad. Sci. USA* 86: 5473-5477), pancreas-specific promoters (Edlund, et al., 1985. *Science* 230: 912-916), and mammary gland-specific promoters (e.g., milk whey promoter; U.S. Pat. No. 4,873,316 and European Application Publication No. 264,166). Developmentally-regulated promoters are also encompassed, e.g., the murine hox promoters (Kessel and Gruss, 1990. *Science* 249: 374-379) and the α-fetoprotein promoter (Campes and Tilghman, 1989. *Genes Dev.* 3: 537-546). With regards to these prokaryotic and eukaryotic vectors, mention is made of U.S. Patent 6,750,059, the contents of which are incorporated by reference herein in their entirety. Other embodiments of the invention may relate to the use of viral vectors, with regards to which mention is made of U.S. Patent application 13/092,085, the contents of which are incorporated by reference herein in their entirety. Tissue-specific regulatory elements are known in the art and in this regard, mention is made of U.S. Patent 7,776,321, the contents of which are incorporated by reference herein in their entirety. In some embodiments, a regulatory element is operably linked to one or more elements of a CRISPR system so as to drive expression of the one or more elements of the CRISPR system.

[00396] In some embodiments, one or more vectors driving expression of one or more elements of a nucleic acid-targeting system are introduced into a host cell such that expression of the elements of the nucleic acid-targeting system direct formation of a nucleic acid-targeting complex at one or more target sites. For example, a nucleic acid-targeting effector enzyme and a nucleic acid-targeting guide RNA could each be operably linked to separate regulatory elements on separate vectors. RNA(s) of the nucleic acid-targeting

system can be delivered to a transgenic nucleic acid-targeting effector protein animal or mammal, e.g., an animal or mammal that constitutively or inducibly or conditionally expresses nucleic acid-targeting effector protein; or an animal or mammal that is otherwise expressing nucleic acid-targeting effector proteins or has cells containing nucleic acid-targeting effector proteins, such as by way of prior administration thereto of a vector or vectors that code for and express in vivo nucleic acid-targeting effector proteins. Alternatively, two or more of the elements expressed from the same or different regulatory elements, may be combined in a single vector, with one or more additional vectors providing any components of the nucleic acid-targeting system not included in the first vector. nucleic acid-targeting system elements that are combined in a single vector may be arranged in any suitable orientation, such as one element located 5' with respect to ("upstream" of) or 3' with respect to ("downstream" of) a second element. The coding sequence of one element may be located on the same or opposite strand of the coding sequence of a second element, and oriented in the same or opposite direction. In some embodiments, a single promoter drives expression of a transcript encoding a nucleic acid-targeting effector protein and the nucleic acid-targeting guide RNA, embedded within one or more intron sequences (e.g., each in a different intron, two or more in at least one intron, or all in a single intron). In some embodiments, the nucleic acid-targeting effector protein and the nucleic acid-targeting guide RNA may be operably linked to and expressed from the same promoter. Delivery vehicles, vectors, particles, nanoparticles, formulations and components thereof for expression of one or more elements of a nucleic acid-targeting system are as used in the foregoing documents, such as WO 2014/093622 (PCT/US2013/074667). In some embodiments, a vector comprises one or more insertion sites, such as a restriction endonuclease recognition sequence (also referred to as a "cloning site"). In some embodiments, one or more insertion sites (e.g., about or more than about 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, or more insertion sites) are located upstream and/or downstream of one or more sequence elements of one or more vectors. When multiple different guide sequences are used, a single expression construct may be used to target nucleic acid-targeting activity to multiple different, corresponding target sequences within a cell. For example, a single vector may comprise about or more than about 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 15, 20, or more guide sequences. In some embodiments, about or more than about 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, or more such guide-sequence-containing vectors may be provided, and optionally delivered to a cell. In some embodiments, a vector comprises a regulatory element operably linked to an enzyme-coding sequence encoding a a nucleic acid-targeting

effector protein. Nucleic acid-targeting effector protein or nucleic acid-targeting guide RNA or RNA(s) can be delivered separately; and advantageously at least one of these is delivered via a particle complex. nucleic acid-targeting effector protein mRNA can be delivered prior to the nucleic acid-targeting guide RNA to give time for nucleic acid-targeting effector protein to be expressed. Nucleic acid-targeting effector protein mRNA might be administered 1-12 hours (preferably around 2-6 hours) prior to the administration of nucleic acid-targeting guide RNA. Alternatively, nucleic acid-targeting effector protein mRNA and nucleic acid-targeting guide RNA can be administered together. Advantageously, a second booster dose of guide RNA can be administered 1-12 hours (preferably around 2-6 hours) after the initial administration of nucleic acid-targeting effector protein mRNA + guide RNA. Additional administrations of nucleic acid-targeting effector protein mRNA and/or guide RNA might be useful to achieve the most efficient levels of genome modification.

[00397] Conventional viral and non-viral based gene transfer methods can be used to introduce nucleic acids in mammalian cells or target tissues. Such methods can be used to administer nucleic acids encoding components of a nucleic acid-targeting system to cells in culture, or in a host organism. Non-viral vector delivery systems include DNA plasmids, RNA (e.g. a transcript of a vector described herein), naked nucleic acid, and nucleic acid complexed with a delivery vehicle, such as a liposome. Viral vector delivery systems include DNA and RNA viruses, which have either episomal or integrated genomes after delivery to the cell. For a review of gene therapy procedures, see Anderson, Science 256:808-813 (1992); Nabel & Felgner, TIBTECH 11:211-217 (1993); Mitani & Caskey, TIBTECH 11:162-166 (1993); Dillon, TIBTECH 11:167-175 (1993); Miller, Nature 357:455-460 (1992); Van Brunt, Biotechnology 6(10):1149-1154 (1988); Vigne, Restorative Neurology and Neuroscience 8:35-36 (1995); Kremer & Perricaudet, British Medical Bulletin 51(1):31-44 (1995); Haddada et al., in Current Topics in Microbiology and Immunology, Doerfler and Böhm (eds) (1995); and Yu et al., Gene Therapy 1:13-26 (1994).

[00398] Methods of non-viral delivery of nucleic acids include lipofection, nucleofection, microinjection, biolistics, virosomes, liposomes, immunoliposomes, polycation or lipid:nucleic acid conjugates, naked DNA, artificial virions, and agent-enhanced uptake of DNA. Lipofection is described in e.g., U.S. Pat. Nos. 5,049,386, 4,946,787; and 4,897,355) and lipofection reagents are sold commercially (e.g., Transfectam™ and Lipofectin™). Cationic and neutral lipids that are suitable for efficient receptor-recognition lipofection of polynucleotides include those of Felgner, WO 91/17424;

WO 91/16024. Delivery can be to cells (e.g. in vitro or ex vivo administration) or target tissues (e.g. in vivo administration).

[00399] Plasmid delivery involves the cloning of a guide RNA into a CRISPR-Cas protein expressing plasmid and transfecting the DNA in cell culture. Plasmid backbones are available commercially and no specific equipment is required. They have the advantage of being modular, capable of carrying different sizes of CRISPR-Cas coding sequences (including those encoding larger sized proteins) as well as selection markers. Both an advantage of plasmids is that they can ensure transient, but sustained expression. However, delivery of plasmids is not straightforward such that in vivo efficiency is often low. The sustained expression can also be disadvantageous in that it can increase off-target editing. In addition excess build-up of the CRISPR-Cas protein can be toxic to the cells. Finally, plasmids always hold the risk of random integration of the dsDNA in the host genome, more particularly in view of the double-stranded breaks being generated (on and off-target).

[00400] The preparation of lipid:nucleic acid complexes, including targeted liposomes such as immunolipid complexes, is well known to one of skill in the art (see, e.g., Crystal, Science 270:404-410 (1995); Blaese et al., Cancer Gene Ther. 2:291-297 (1995); Behr et al., Bioconjugate Chem. 5:382-389 (1994); Remy et al., Bioconjugate Chem. 5:647-654 (1994); Gao et al., Gene Therapy 2:710-722 (1995); Ahmad et al., Cancer Res. 52:4817-4820 (1992); U.S. Pat. Nos. 4,186,183, 4,217,344, 4,235,871, 4,261,975, 4,485,054, 4,501,728, 4,774,085, 4,837,028, and 4,946,787). This is discussed more in detail below.

[00401] The use of RNA or DNA viral based systems for the delivery of nucleic acids takes advantage of highly evolved processes for targeting a virus to specific cells in the body and trafficking the viral payload to the nucleus. Viral vectors can be administered directly to patients (in vivo) or they can be used to treat cells in vitro, and the modified cells may optionally be administered to patients (ex vivo). Conventional viral based systems could include retroviral, lentivirus, adenoviral, adeno-associated and herpes simplex virus vectors for gene transfer. Integration in the host genome is possible with the retrovirus, lentivirus, and adeno-associated virus gene transfer methods, often resulting in long term expression of the inserted transgene. Additionally, high transduction efficiencies have been observed in many different cell types and target tissues.

[00402] The tropism of a retrovirus can be altered by incorporating foreign envelope proteins, expanding the potential target population of target cells. Lentiviral vectors are retroviral vectors that are able to transduce or infect non-dividing cells and typically produce high viral titers. Selection of a retroviral gene transfer system would therefore

depend on the target tissue. Retroviral vectors are comprised of cis-acting long terminal repeats with packaging capacity for up to 6-10 kb of foreign sequence. The minimum cis-acting LTRs are sufficient for replication and packaging of the vectors, which are then used to integrate the therapeutic gene into the target cell to provide permanent transgene expression. Widely used retroviral vectors include those based upon murine leukemia virus (MuLV), gibbon ape leukemia virus (GaLV), Simian Immuno deficiency virus (SIV), human immuno deficiency virus (HIV), and combinations thereof (see, e.g., Buchscher et al., J. Virol. 66:2731-2739 (1992); Johann et al., J. Virol. 66:1635-1640 (1992); Sommnerfelt et al., Virol. 176:58-59 (1990); Wilson et al., J. Virol. 63:2374-2378 (1989); Miller et al., J. Virol. 65:2220-2224 (1991); PCT/US94/05700).

[00403] In applications where transient expression is preferred, adenoviral based systems may be used. Adenoviral based vectors are capable of very high transduction efficiency in many cell types and do not require cell division. With such vectors, high titer and levels of expression have been obtained. This vector can be produced in large quantities in a relatively simple system. Adeno-associated virus ("AAV") vectors may also be used to transduce cells with target nucleic acids, e.g., in the in vitro production of nucleic acids and peptides, and for in vivo and ex vivo gene therapy procedures (see, e.g., West et al., Virology 160:38-47 (1987); U.S. Pat. No. 4,797,368; WO 93/24641; Kotin, Human Gene Therapy 5:793-801 (1994); Muzyczka, J. Clin. Invest. 94:1351 (1994). Construction of recombinant AAV vectors are described in a number of publications, including U.S. Pat. No. 5,173,414; Tratschin et al., Mol. Cell. Biol. 5:3251-3260 (1985); Tratschin, et al., Mol. Cell. Biol. 4:2072-2081 (1984); Hermonat & Muzyczka, PNAS 81:6466-6470 (1984); and Samulski et al., J. Virol. 63:03822-3828 (1989).

[00404] The invention provides AAV that contains or consists essentially of an exogenous nucleic acid molecule encoding a CRISPR system, e.g., a plurality of cassettes comprising or consisting a first cassette comprising or consisting essentially of a promoter, a nucleic acid molecule encoding a CRISPR-associated (Cas) protein (putative nuclease or helicase proteins), e.g., Cpf1 and a terminator, andone or more, advantageously up to the packaging size limit of the vector, e.g., in total (including the first cassette) five, cassettes comprising or consisting essentially of a promoter, nucleic acid molecule encoding guide RNA (gRNA) and a terminator (e.g., each cassette schematically represented as Promoter-gRNA1-terminator, Promoter-gRNA2-terminator ... Promoter-gRNA(N)-terminator, where N is a number that can be inserted that is at an upper limit of the packaging size limit of the vector), or two or more individual rAAVs, each containing one or more than one cassette of

a CRISPR system, e.g., a first rAAV containing the first cassette comprising or consisting essentially of a promoter, a nucleic acid molecule encoding Cas, e.g., Cas (Cpf1) and a terminator, and a second rAAV containing one or more cassettes each comprising or consisting essentially of a promoter, nucleic acid molecule encoding guide RNA (gRNA) and a terminator (e.g., each cassette schematically represented as Promoter-gRNA1-terminator, Promoter-gRNA2-terminator ... Promoter-gRNA(N)-terminator, where N is a number that can be inserted that is at an upper limit of the packaging size limit of the vector). Alternatively, because Cpf1 can process its own crRNA/gRNA, a single crRNA/gRNA array can be used for multiplex gene editing. Hence, instead of including multiple cassettes to deliver the gRNAs, the rAAV may contain a single cassette comprising or consisting essentially of a promoter, a plurality of crRNA/gRNA, and a terminator (e.g., schematically represented as Promoter-gRNA1-gRNA2 ...gRNA(N)-terminator, where N is a number that can be inserted that is at an upper limit of the packaging size limit of the vector). See Zetsche *et al Nature Biotechnology* 35, 31–34 (2017), which is incorporated herein by reference in its entirety. As rAAV is a DNA virus, the nucleic acid molecules in the herein discussion concerning AAV or rAAV are advantageously DNA. The promoter is in some embodiments advantageously human Synapsin I promoter (hSyn). Additional methods for the delivery of nucleic acids to cells are known to those skilled in the art. See, for example, US20030087817, incorporated herein by reference.

[00405] In another embodiment, Cocal vesiculovirus envelope pseudotyped retroviral vector particles are contemplated (see, e.g., US Patent Publication No. 20120164118 assigned to the Fred Hutchinson Cancer Research Center). Cocal virus is in the Vesiculovirus genus, and is a causative agent of vesicular stomatitis in mammals. Cocal virus was originally isolated from mites in Trinidad (Jonkers et al., Am. J. Vet. Res. 25:236-242 (1964)), and infections have been identified in Trinidad, Brazil, and Argentina from insects, cattle, and horses. Many of the vesiculoviruses that infect mammals have been isolated from naturally infected arthropods, suggesting that they are vector-borne. Antibodies to vesiculoviruses are common among people living in rural areas where the viruses are endemic and laboratory-acquired; infections in humans usually result in influenza-like symptoms. The Cocal virus envelope glycoprotein shares 71.5% identity at the amino acid level with VSV-G Indiana, and phylogenetic comparison of the envelope gene of vesiculoviruses shows that Cocal virus is serologically distinct from, but most closely related to, VSV-G Indiana strains among the vesiculoviruses. Jonkers et al., Am. J. Vet. Res. 25:236-242 (1964) and Travassos da Rosa et al., Am. J. Tropical Med. & Hygiene

33:999-1006 (1984). The Cocal vesiculovirus envelope pseudotyped retroviral vector particles may include for example, lentiviral, alpharetroviral, betaretroviral, gammaretroviral, deltaretroviral, and epsilonretroviral vector particles that may comprise retroviral Gag, Pol, and/or one or more accessory protein(s) and a Cocal vesiculovirus envelope protein. Within certain aspects of these embodiments, the Gag, Pol, and accessory proteins are lentiviral and/or gammaretroviral.

[00406] In some embodiments, a host cell is transiently or non-transiently transfected with one or more vectors described herein. In some embodiments, a cell is transfected as it naturally occurs in a subject optionally to be reintroduced therein. In some embodiments, a cell that is transfected is taken from a subject. In some embodiments, the cell is derived from cells taken from a subject, such as a cell line. A wide variety of cell lines for tissue culture are known in the art. Examples of cell lines include, but are not limited to, C8161, CCRF-CEM, MOLT, mIMCD-3, NHDF, HeLa-S3, Huh1, Huh4, Huh7, HUVEC, HASMC, HEKn, HEKa, MiaPaCell, Panc1, PC-3, TF1, CTLL-2, C1R, Rat6, CV1, RPTE, A10, T24, J82, A375, ARH-77, Calu1, SW480, SW620, SKOV3, SK-UT, CaCo2, P388D1, SEM-K2, WEHI-231, HB56, TIB55, Jurkat, J45.01, LRMB, Bcl-1, BC-3, IC21, DLD2, Raw264.7, NRK, NRK-52E, MRC5, MEF, Hep G2, HeLa B, HeLa T4, COS, COS-1, COS-6, COS-M6A, BS-C-1 monkey kidney epithelial, BALB/ 3T3 mouse embryo fibroblast, 3T3 Swiss, 3T3-L1, 132-d5 human fetal fibroblasts; 10.1 mouse fibroblasts, 293-T, 3T3, 721, 9L, A2780, A2780ADR, A2780cis, A172, A20, A253, A431, A-549, ALC, B16, B35, BCP-1 cells, BEAS-2B, bEnd.3, BHK-21, BR 293, BxPC3, C3H-10T1/2, C6/36, Cal-27, CHO, CHO-7, CHO-IR, CHO-K1, CHO-K2, CHO-T, CHO Dhfr -/-, COR-L23, COR-L23/CPR, COR-L23/5010, COR-L23/R23, COS-7, COV-434, CML T1, CMT, CT26, D17, DH82, DU145, DuCaP, EL4, EM2, EM3, EMT6/AR1, EMT6/AR10.0, FM3, H1299, H69, HB54, HB55, HCA2, HEK-293, HeLa, Hepa1c1c7, HL-60, HMEC, HT-29, Jurkat, JY cells, K562 cells, Ku812, KCL22, KG1, KYO1, LNCap, Ma-Mel 1-48, MC-38, MCF-7, MCF-10A, MDA-MB-231, MDA-MB-468, MDA-MB-435, MDCK II, MDCK II, MOR/0.2R, MONO-MAC 6, MTD-1A, MyEnd, NCI-H69/CPR, NCI-H69/LX10, NCI-H69/LX20, NCI-H69/LX4, NIH-3T3, NALM-1, NW-145, OPCN / OPCT cell lines, Peer, PNT-1A / PNT 2, RenCa, RIN-5F, RMA/RMAS, Saos-2 cells, Sf-9, SkBr3, T2, T-47D, T84, THP1 cell line, U373, U87, U937, VCaP, Vero cells, WM39, WT-49, X63, YAC-1, YAR, and transgenic varieties thereof. Cell lines are available from a variety of sources known to those with skill in the art (see, e.g., the American Type Culture Collection (ATCC) (Manassus, Va.)).

[00407] In particular embodiments, transient expression and/or presence of one or more of the components of the AD-functionalized CRISPR system can be of interest, such as to reduce off-target effects. In some embodiments, a cell transfected with one or more vectors described herein is used to establish a new cell line comprising one or more vector-derived sequences. In some embodiments, a cell transiently transfected with the components of a AD-functionalized CRISPR system as described herein (such as by transient transfection of one or more vectors, or transfection with RNA), and modified through the activity of a CRISPR complex, is used to establish a new cell line comprising cells containing the modification but lacking any other exogenous sequence. In some embodiments, cells transiently or non-transiently transfected with one or more vectors described herein, or cell lines derived from such cells are used in assessing one or more test compounds.

[00408] In some embodiments it is envisaged to introduce the RNA and/or protein directly to the host cell. For instance, the CRISPR-Cas protein can be delivered as encoding mRNA together with an in vitro transcribed guide RNA. Such methods can reduce the time to ensure effect of the CRISPR-Cas protein and further prevents long-term expression of the CRISPR system components.

[00409] In some embodiments the RNA molecules of the invention are delivered in liposome or lipofectin formulations and the like and can be prepared by methods well known to those skilled in the art. Such methods are described, for example, in U.S. Pat. Nos. 5,593,972, 5,589,466, and 5,580,859, which are herein incorporated by reference. Delivery systems aimed specifically at the enhanced and improved delivery of siRNA into mammalian cells have been developed, (see, for example, Shen et al FEBS Let. 2003, 539:111-114; Xia et al., Nat. Biotech. 2002, 20:1006-1010; Reich et al., Mol. Vision. 2003, 9: 210-216; Sorensen et al., J. Mol. Biol. 2003, 327: 761-766; Lewis et al., Nat. Gen. 2002, 32: 107-108 and Simeoni et al., NAR 2003, 31, 11: 2717-2724) and may be applied to the present invention. siRNA has recently been successfully used for inhibition of gene expression in primates (see for example. Tolentino et al., Retina 24(4):660 which may also be applied to the present invention.

[00410] Indeed, RNA delivery is a useful method of in vivo delivery. It is possible to deliver Cpf1, adenosine deaminase, and guide RNA into cells using liposomes or nanoparticles. Thus delivery of the CRISPR-Cas protein, such as a Cpf1, the delivery of the adenosine deaminase (which may be fused to the CRISPR-Cas protein or an adaptor protein), and/or delivery of the RNAs of the invention may be in RNA form and via microvesicles, liposomes or particle or particles. For example, Cpf1 mRNA, adenosine

deaminase mRNA, and guide RNA can be packaged into liposomal particles for delivery in vivo. Liposomal transfection reagents such as lipofectamine from Life Technologies and other reagents on the market can effectively deliver RNA molecules into the liver.

[00411] Means of delivery of RNA also preferred include delivery of RNA via particles (Cho, S., Goldberg, M., Son, S., Xu, Q., Yang, F., Mei, Y., Bogatyrev, S., Langer, R. and Anderson, D., Lipid-like nanoparticles for small interfering RNA delivery to endothelial cells, Advanced Functional Materials, 19: 3112-3118, 2010) or exosomes (Schroeder, A., Levins, C., Cortez, C., Langer, R., and Anderson, D., Lipid-based nanotherapeutics for siRNA delivery, Journal of Internal Medicine, 267: 9-21, 2010, PMID: 20059641). Indeed, exosomes have been shown to be particularly useful in delivery siRNA, a system with some parallels to the CRISPR system. For instance, El-Andaloussi S, et al. ("Exosome-mediated delivery of siRNA in vitro and in vivo." Nat Protoc. 2012 Dec;7(12):2112-26. doi: 10.1038/nprot.2012.131. Epub 2012 Nov 15.) describe how exosomes are promising tools for drug delivery across different biological barriers and can be harnessed for delivery of siRNA in vitro and in vivo. Their approach is to generate targeted exosomes through transfection of an expression vector, comprising an exosomal protein fused with a peptide ligand. The exosomes are then purify and characterized from transfected cell supernatant, then RNA is loaded into the exosomes. Delivery or administration according to the invention can be performed with exosomes, in particular but not limited to the brain. Vitamin E (α-tocopherol) may be conjugated with CRISPR Cas and delivered to the brain along with high density lipoprotein (HDL), for example in a similar manner as was done by Uno et al. (HUMAN GENE THERAPY 22:711–719 (June 2011)) for delivering short-interfering RNA (siRNA) to the brain. Mice were infused via Osmotic minipumps (model 1007D; Alzet, Cupertino, CA) filled with phosphate-buffered saline (PBS) or free TocsiBACE or Toc-siBACE/HDL and connected with Brain Infusion Kit 3 (Alzet). A brain-infusion cannula was placed about 0.5mm posterior to the bregma at midline for infusion into the dorsal third ventricle. Uno et al. found that as little as 3 nmol of Toc-siRNA with HDL could induce a target reduction in comparable degree by the same ICV infusion method. A similar dosage of CRISPR Cas conjugated to α-tocopherol and co-administered with HDL targeted to the brain may be contemplated for humans in the present invention, for example, about 3 nmol to about 3 μmol of CRISPR Cas targeted to the brain may be contemplated. Zou et al. ((HUMAN GENE THERAPY 22:465-475 (April 2011)) describes a method of lentiviral-mediated delivery of short-hairpin RNAs targeting PKCγ for in vivo gene silencing in the spinal cord of rats. Zou et al. administered about 10 μl of a

recombinant lentivirus having a titer of 1 x 10^9 transducing units (TU)/ml by an intrathecal catheter. A similar dosage of CRISPR Cas expressed in a lentiviral vector targeted to the brain may be contemplated for humans in the present invention, for example, about 10-50 ml of CRISPR Cas targeted to the brain in a lentivirus having a titer of 1 x 10^9 transducing units (TU)/ml may be contemplated.

[00412] Dosage of vectors

[00413] In some embodiments, the vector, e.g., plasmid or viral vector is delivered to the tissue of interest by, for example, an intramuscular injection, while other times the delivery is via intravenous, transdermal, intranasal, oral, mucosal, or other delivery methods. Such delivery may be either via a single dose, or multiple doses. One skilled in the art understands that the actual dosage to be delivered herein may vary greatly depending upon a variety of factors, such as the vector choice, the target cell, organism, or tissue, the general condition of the subject to be treated, the degree of transformation/modification sought, the administration route, the administration mode, the type of transformation/modification sought, etc.

[00414] Such a dosage may further contain, for example, a carrier (water, saline, ethanol, glycerol, lactose, sucrose, calcium phosphate, gelatin, dextran, agar, pectin, peanut oil, sesame oil, etc.), a diluent, a pharmaceutically-acceptable carrier (e.g., phosphate-buffered saline), a pharmaceutically-acceptable excipient, and/or other compounds known in the art. The dosage may further contain one or more pharmaceutically acceptable salts such as, for example, a mineral acid salt such as a hydrochloride, a hydrobromide, a phosphate, a sulfate, etc.; and the salts of organic acids such as acetates, propionates, malonates, benzoates, etc. Additionally, auxiliary substances, such as wetting or emulsifying agents, pH buffering substances, gels or gelling materials, flavorings, colorants, microspheres, polymers, suspension agents, etc. may also be present herein. In addition, one or more other conventional pharmaceutical ingredients, such as preservatives, humectants, suspending agents, surfactants, antioxidants, anticaking agents, fillers, chelating agents, coating agents, chemical stabilizers, etc. may also be present, especially if the dosage form is a reconstitutable form. Suitable exemplary ingredients include microcrystalline cellulose, carboxymethylcellulose sodium, polysorbate 80, phenylethyl alcohol, chlorobutanol, potassium sorbate, sorbic acid, sulfur dioxide, propyl gallate, the parabens, ethyl vanillin, glycerin, phenol, parachlorophenol, gelatin, albumin and a combination thereof. A thorough discussion of pharmaceutically acceptable excipients is available in REMINGTON'S

PHARMACEUTICAL SCIENCES (Mack Pub. Co., N.J. 1991) which is incorporated by reference herein.

[00415] In an embodiment herein the delivery is via an adenovirus, which may be at a single booster dose containing at least 1 x 10^5 particles (also referred to as particle units, pu) of adenoviral vector. In an embodiment herein, the dose preferably is at least about 1 x 10^6 particles (for example, about 1 x 10^6-1 x 10^{12} particles), more preferably at least about 1 x 10^7 particles, more preferably at least about 1 x 10^8 particles (e.g., about 1 x 10^8-1 x 10^{11} particles or about 1 x 10^8-1 x 10^{12} particles), and most preferably at least about 1 x 10^0 particles (e.g., about 1 x 10^9-1 x 10^{10} particles or about 1 x 10^9-1 x 10^{12} particles), or even at least about 1 x 10^{10} particles (e.g., about 1 x 10^{10}-1 x 10^{12} particles) of the adenoviral vector. Alternatively, the dose comprises no more than about 1 x 10^{14} particles, preferably no more than about 1 x 10^{13} particles, even more preferably no more than about 1 x 10^{12} particles, even more preferably no more than about 1 x 10^{11} particles, and most preferably no more than about 1 x 10^{10} particles (e.g., no more than about 1 x 10^9 articles). Thus, the dose may contain a single dose of adenoviral vector with, for example, about 1 x 10^6 particle units (pu), about 2 x 10^6 pu, about 4 x 10^6 pu, about 1 x 10^7 pu, about 2 x 10^7 pu, about 4 x 10^7 pu, about 1 x 10^8 pu, about 2 x 10^8 pu, about 4 x 10^8 pu, about 1 x 10^9 pu, about 2 x 10^9 pu, about 4 x 10^9 pu, about 1 x 10^{10} pu, about 2 x 10^{10} pu, about 4 x 10^{10} pu, about 1 x 10^{11} pu, about 2 x 10^{11} pu, about 4 x 10^{11} pu, about 1 x 10^{12} pu, about 2 x 10^{12} pu, or about 4 x 10^{12} pu of adenoviral vector. See, for example, the adenoviral vectors in U.S. Patent No. 8,454,972 B2 to Nabel, et. al., granted on June 4, 2013; incorporated by reference herein, and the dosages at col 29, lines 36-58 thereof. In an embodiment herein, the adenovirus is delivered via multiple doses.

[00416] In an embodiment herein, the delivery is via an AAV. A therapeutically effective dosage for in vivo delivery of the AAV to a human is believed to be in the range of from about 20 to about 50 ml of saline solution containing from about 1 x 10^{10} to about 1 x 10^{10} functional AAV/ml solution. The dosage may be adjusted to balance the therapeutic benefit against any side effects. In an embodiment herein, the AAV dose is generally in the range of concentrations of from about 1 x 10^5 to 1 x 10^{50} genomes AAV, from about 1 x 10^8 to 1 x 10^{20} genomes AAV, from about 1 x 10^{10} to about 1 x 10^{16} genomes, or about 1 x 10^{11} to about 1 x 10^{16} genomes AAV. A human dosage may be about 1 x 10^{13} genomes AAV. Such concentrations may be delivered in from about 0.001 ml to about 100 ml, about 0.05 to about 50 ml, or about 10 to about 25 ml of a carrier solution. Other effective dosages can be readily established by one of ordinary skill in the art through routine trials establishing

dose response curves. See, for example, U.S. Patent No. 8,404,658 B2 to Hajjar, et al., granted on March 26, 2013, at col. 27, lines 45-60.

[00417] In an embodiment herein the delivery is via a plasmid. In such plasmid compositions, the dosage should be a sufficient amount of plasmid to elicit a response. For instance, suitable quantities of plasmid DNA in plasmid compositions can be from about 0.1 to about 2 mg, or from about 1 μg to about 10 μg per 70 kg individual. Plasmids of the invention will generally comprise (i) a promoter; (ii) a sequence encoding a CRISPR-Cas protein, operably linked to said promoter; (iii) a selectable marker; (iv) an origin of replication; and (v) a transcription terminator downstream of and operably linked to (ii). The plasmid can also encode the RNA components of a CRISPR complex, but one or more of these may instead be encoded on a different vector.

[00418] The doses herein are based on an average 70 kg individual. The frequency of administration is within the ambit of the medical or veterinary practitioner (e.g., physician, veterinarian), or scientist skilled in the art. It is also noted that mice used in experiments are typically about 20g and from mice experiments one can scale up to a 70 kg individual.

[00419] The dosage used for the compositions provided herein include dosages for repeated administration or repeat dosing. In particular embodiments, the administration is repeated within a period of several weeks, months, or years. Suitable assays can be performed to obtain an optimal dosage regime. Repeated administration can allow the use of lower dosage, which can positively affect off-target modifications.

[00420] RNA delivery

[00421] In particular embodiments, RNA based delivery is used. In these embodiments, mRNA of the CRISPR-Cas protein, mRNA of the adenosine deaminase (which may be fused to a CRISPR-Cas protein or an adaptor), are delivered together with in vitro transcribed guide RNA. Liang et al. describes efficient genome editing using RNA based delivery (Protein Cell. 2015 May; 6(5): 363–372). In some embodiments, the mRNA(s) encoding Cpf1 and/or adenosine deaminase can be chemically modified, which may lead to improved activity compared to plasmid-encoded Cpf1 and/or adenosine deaminase. For example, uridines in the mRNA(s) can be partially or fully substituted with pseudouridine (Ψ), N^1-methylpseudouridine ($me^1\Psi$), 5-methoxyuridine(5moU). See Li *et al.*, *Nature Biomedical Engineering* 1, 0066 DOI:10.1038/s41551-017-0066 (2017), which is incorporated herein by reference in its entirety.

[00422] RNP

[00423] In particular embodiments, pre-complexed guide RNA, CRISPR-Cas protein, and adenosine deaminase (which may be fused to a CRISPR-Cas protein or an adaptor) are delived as a ribonucleoprotein (RNP). RNPs have the advantage that they lead to rapid editing effects even more so than the RNA method because this process avoids the need for transcription. An important advantage is that both RNP delivery is transient, reducing off-target effects and toxicity issues. Efficient genome editing in different cell types has been observed by Kim et al. (2014, Genome Res. 24(6):1012-9), Paix et al. (2015, Genetics 204(1):47-54), Chu et al. (2016, BMC Biotechnol. 16:4), and Wang et al. (2013, Cell. 9;153(4):910-8).

[00424] In particular embodiments, the ribonucleoprotein is delivered by way of a polypeptide-based shuttle agent as described in WO2016161516. WO2016161516 describes efficient transduction of polypeptide cargos using synthetic peptides comprising an endosome leakage domain (ELD) operably linked to a cell penetrating domain (CPD), to a histidine-rich domain and a CPD. Similarly these polypeptides can be used for the delivery of CRISPR-effector based RNPs in eukaryotic cells

[00425] Particles

[00426] In some aspects or embodiments, a composition comprising a delivery particle formulation may be used. In some aspects or embodiments, the formulation comprises a CRISPR complex, the complex comprising a CRISPR protein and a guide which directs sequence-specific binding of the CRISPR complex to a target sequence. In some embodiments, the delivery particle comprises a lipid-based particle, optionally a lipid nanoparticle, or cationic lipid and optionally biodegradable polymer. In some embodiments, the cationic lipid comprises 1,2-dioleoyl-3-trimethylammonium-propane (DOTAP). In some embodiments, the hydrophilic polymer comprises ethylene glycol or polyethylene glycol. In some embodiments, the delivery particle further comprises a lipoprotein, preferably cholesterol. In some embodiments, the delivery particles are less than 500 nm in diameter, optionally less than 250 nm in diameter, optionally less than 100 nm in diameter, optionally about 35 nm to about 60 nm in diameter.

[00427] Several types of particle delivery systems and/or formulations are known to be useful in a diverse spectrum of biomedical applications. In general, a particle is defined as a small object that behaves as a whole unit with respect to its transport and properties. Particles are further classified according to diameter. Coarse particles cover a range between 2,500 and 10,000 nanometers. Fine particles are sized between 100 and 2,500 nanometers. Ultrafine particles, or nanoparticles, are generally between 1 and 100

nanometers in size. The basis of the 100-nm limit is the fact that novel properties that differentiate particles from the bulk material typically develop at a critical length scale of under 100 nm.

[00428] As used herein, a particle delivery system/formulation is defined as any biological delivery system/formulation which includes a particle in accordance with the present invention. A particle in accordance with the present invention is any entity having a greatest dimension (e.g. diameter) of less than 100 microns (μm). In some embodiments, inventive particles have a greatest dimension of less than 10 μ m. In some embodiments, inventive particles have a greatest dimension of less than 2000 nanometers (nm). In some embodiments, inventive particles have a greatest dimension of less than 1000 nanometers (nm). In some embodiments, inventive particles have a greatest dimension of less than 900 nm, 800 nm, 700 nm, 600 nm, 500 nm, 400 nm, 300 nm, 200 nm, or 100 nm. Typically, inventive particles have a greatest dimension (e.g., diameter) of 500 nm or less. In some embodiments, inventive particles have a greatest dimension (e.g., diameter) of 250 nm or less. In some embodiments, inventive particles have a greatest dimension (e.g., diameter) of 200 nm or less. In some embodiments, inventive particles have a greatest dimension (e.g., diameter) of 150 nm or less. In some embodiments, inventive particles have a greatest dimension (e.g., diameter) of 100 nm or less. Smaller particles, e.g., having a greatest dimension of 50 nm or less are used in some embodiments of the invention. In some embodiments, inventive particles have a greatest dimension ranging between 25 nm and 200 nm.

[00429] In terms of this invention, it is preferred to have one or more components of CRISPR complex, e.g., CRISPR-Cas protein or mRNA, or adenosine deaminase (which may be fused to a CRISPR-Cas protein or an adaptor) or mRNA, or guide RNA delivered using nanoparticles or lipid envelopes. Other delivery systems or vectors are may be used in conjunction with the nanoparticle aspects of the invention.

[00430] In general, a "nanoparticle" refers to any particle having a diameter of less than 1000 nm. In certain preferred embodiments, nanoparticles of the invention have a greatest dimension (e.g., diameter) of 500 nm or less. In other preferred embodiments, nanoparticles of the invention have a greatest dimension ranging between 25 nm and 200 nm. In other preferred embodiments, nanoparticles of the invention have a greatest dimension of 100 nm or less. In other preferred embodiments, nanoparticles of the invention have a greatest dimension ranging between 35 nm and 60 nm. It will be appreciated that reference made herein to particles or nanoparticles can be interchangeable, where appropriate.

[00431] It will be understood that the size of the particle will differ depending as to whether it is measured before or after loading. Accordingly, in particular embodiments, the term "nanoparticles" may apply only to the particles pre loading.

[00432] Nanoparticles encompassed in the present invention may be provided in different forms, e.g., as solid nanoparticles (e.g., metal such as silver, gold, iron, titanium), non-metal, lipid-based solids, polymers), suspensions of nanoparticles, or combinations thereof. Metal, dielectric, and semiconductor nanoparticles may be prepared, as well as hybrid structures (e.g., core–shell nanoparticles). Nanoparticles made of semiconducting material may also be labeled quantum dots if they are small enough (typically sub 10 nm) that quantization of electronic energy levels occurs. Such nanoscale particles are used in biomedical applications as drug carriers or imaging agents and may be adapted for similar purposes in the present invention.

[00433] Semi-solid and soft nanoparticles have been manufactured, and are within the scope of the present invention. A prototype nanoparticle of semi-solid nature is the liposome. Various types of liposome nanoparticles are currently used clinically as delivery systems for anticancer drugs and vaccines. Nanoparticles with one half hydrophilic and the other half hydrophobic are termed Janus particles and are particularly effective for stabilizing emulsions. They can self-assemble at water/oil interfaces and act as solid surfactants.

[00434] Particle characterization (including e.g., characterizing morphology, dimension, etc.) is done using a variety of different techniques. Common techniques are electron microscopy (TEM, SEM), atomic force microscopy (AFM), dynamic light scattering (DLS), X-ray photoelectron spectroscopy (XPS), powder X-ray diffraction (XRD), Fourier transform infrared spectroscopy (FTIR), matrix-assisted laser desorption/ionization time-of-flight mass spectrometry(MALDI-TOF), ultraviolet-visible spectroscopy, dual polarization interferometry and nuclear magnetic resonance (NMR). Characterization (dimension measurements) may be made as to native particles (i.e., preloading) or after loading of the cargo (herein cargo refers to e.g., one or more components of CRISPR-Cas system e.g., CRISPR-Cas protein or mRNA, adenosine deaminase (which may be fused to a CRISPR-Cas protein or an adaptor) or mRNA, or guide RNA, or any combination thereof, and may include additional carriers and/or excipients) to provide particles of an optimal size for delivery for any *in vitro*, *ex vivo* and/or *in vivo* application of the present invention. In certain preferred embodiments, particle dimension (e.g., diameter) characterization is based on measurements using dynamic laser scattering (DLS). Mention is made of US Patent No.

8,709,843; US Patent No. 6,007,845; US Patent No. 5,855,913; US Patent No. 5,985,309; US. Patent No. 5,543,158; and the publication by James E. Dahlman and Carmen Barnes et al. Nature Nanotechnology (2014) published online 11 May 2014, doi:10.1038/nnano.2014.84, concerning particles, methods of making and using them and measurements thereof.

[00435] Particles delivery systems within the scope of the present invention may be provided in any form, including but not limited to solid, semi-solid, emulsion, or colloidal particles. As such any of the delivery systems described herein, including but not limited to, e.g., lipid-based systems, liposomes, micelles, microvesicles, exosomes, or gene gun may be provided as particle delivery systems within the scope of the present invention.

[00436] CRISPR-Cas protein mRNA, adenosine deaminase (which may be fused to a CRISPR-Cas protein or an adaptor) or mRNA, and guide RNA may be delivered simultaneously using particles or lipid envelopes; for instance, CRISPR-Cas protein and RNA of the invention, e.g., as a complex, can be delivered via a particle as in Dahlman et al., WO2015089419 A2 and documents cited therein, such as 7C1 (see, e.g., James E. Dahlman and Carmen Barnes et al. Nature Nanotechnology (2014) published online 11 May 2014, doi:10.1038/nnano.2014.84), e.g., delivery particle comprising lipid or lipidoid and hydrophilic polymer, e.g., cationic lipid and hydrophilic polymer, for instance wherein the cationic lipid comprises 1,2-dioleoyl-3-trimethylammonium-propane (DOTAP) or 1,2-ditetradecanoyl-*sn*-glycero-3-phosphocholine (DMPC) and/or wherein the hydrophilic polymer comprises ethylene glycol or polyethylene glycol (PEG); and/or wherein the particle further comprises cholesterol (e.g., particle from formulation 1 = DOTAP 100, DMPC 0, PEG 0, Cholesterol 0; formulation number 2 = DOTAP 90, DMPC 0, PEG 10, Cholesterol 0; formulation number 3 = DOTAP 90, DMPC 0, PEG 5, Cholesterol 5), wherein particles are formed using an efficient, multistep process wherein first, effector protein and RNA are mixed together, e.g., at a 1:1 molar ratio, e.g., at room temperature, e.g., for 30 minutes, e.g., in sterile, nuclease free 1X PBS; and separately, DOTAP, DMPC, PEG, and cholesterol as applicable for the formulation are dissolved in alcohol, e.g., 100% ethanol; and, the two solutions are mixed together to form particles containing the complexes).

[00437] Nucleic acid-targeting effector proteins (e.g., a Type V protein such as Cpf1) mRNA and guide RNA may be delivered simultaneously using particles or lipid envelopes. Examples of suitable particles include but are not limited to those described in US 9,301,923.

[00438] For example, Su X, Fricke J, Kavanagh DG, Irvine DJ ("In vitro and in vivo mRNA delivery using lipid-enveloped pH-responsive polymer nanoparticles" Mol Pharm. 2011 Jun 6;8(3):774-87. doi: 10.1021/mp100390w. Epub 2011 Apr 1) describes biodegradable core-shell structured nanoparticles with a poly(β-amino ester) (PBAE) core enveloped by a phospholipid bilayer shell. These were developed for in vivo mRNA delivery. The pH-responsive PBAE component was chosen to promote endosome disruption, while the lipid surface layer was selected to minimize toxicity of the polycation core. Such are, therefore, preferred for delivering RNA of the present invention.

[00439] In one embodiment, particles/nanoparticles based on self assembling bioadhesive polymers are contemplated, which may be applied to oral delivery of peptides, intravenous delivery of peptides and nasal delivery of peptides, all to the brain. Other embodiments, such as oral absorption and ocular delivery of hydrophobic drugs are also contemplated. The molecular envelope technology involves an engineered polymer envelope which is protected and delivered to the site of the disease (see, e.g., Mazza, M. et al. ACSNano, 2013. 7(2): 1016-1026; Siew, A., et al. Mol Pharm, 2012. 9(1):14-28; Lalatsa, A., et al. J Contr Rel, 2012. 161(2):523-36; Lalatsa, A., et al., Mol Pharm, 2012. 9(6):1665-80; Lalatsa, A., et al. Mol Pharm, 2012. 9(6):1764-74; Garrett, N.L., et al. J Biophotonics, 2012. 5(5-6):458-68; Garrett, N.L., et al. J Raman Spect, 2012. 43(5):681-688; Ahmad, S., et al. J Royal Soc Interface 2010. 7:S423-33; Uchegbu, I.F. Expert Opin Drug Deliv, 2006. 3(5):629-40; Qu, X.,et al. Biomacromolecules, 2006. 7(12):3452-9 and Uchegbu, I.F., et al. Int J Pharm, 2001. 224:185-199). Doses of about 5 mg/kg are contemplated, with single or multiple doses, depending on the target tissue.

[00440] In one embodiment, particles/nanoparticles that can deliver RNA to a cancer cell to stop tumor growth developed by Dan Anderson's lab at MIT may be used/and or adapted to the AD-functionalized CRISPR-Cas system of the present invention. In particular, the Anderson lab developed fully automated, combinatorial systems for the synthesis, purification, characterization, and formulation of new biomaterials and nanoformulations. See, e.g., Alabi et al., Proc Natl Acad Sci U S A. 2013 Aug 6;110(32):12881-6; Zhang et al., Adv Mater. 2013 Sep 6;25(33):4641-5; Jiang et al., Nano Lett. 2013 Mar 13;13(3):1059-64; Karagiannis et al., ACS Nano. 2012 Oct 23;6(10):8484-7; Whitehead et al., ACS Nano. 2012 Aug 28;6(8):6922-9 and Lee et al., Nat Nanotechnol. 2012 Jun 3;7(6):389-93.

[00441] US patent application 20110293703 relates to lipidoid compounds are also particularly useful in the administration of polynucleotides, which may be applied to deliver the AD-functionalized CRISPR-Cas system of the present invention. In one aspect, the

aminoalcohol lipidoid compounds are combined with an agent to be delivered to a cell or a subject to form microparticles, nanoparticles, liposomes, or micelles. The agent to be delivered by the particles, liposomes, or micelles may be in the form of a gas, liquid, or solid, and the agent may be a polynucleotide, protein, peptide, or small molecule. The aminoalcohol lipidoid compounds may be combined with other aminoalcohol lipidoid compounds, polymers (synthetic or natural), surfactants, cholesterol, carbohydrates, proteins, lipids, etc. to form the particles. These particles may then optionally be combined with a pharmaceutical excipient to form a pharmaceutical composition.

[00442] US Patent Publication No. 20110293703 also provides methods of preparing the aminoalcohol lipidoid compounds. One or more equivalents of an amine are allowed to react with one or more equivalents of an epoxide-terminated compound under suitable conditions to form an aminoalcohol lipidoid compound of the present invention. In certain embodiments, all the amino groups of the amine are fully reacted with the epoxide-terminated compound to form tertiary amines. In other embodiments, all the amino groups of the amine are not fully reacted with the epoxide-terminated compound to form tertiary amines thereby resulting in primary or secondary amines in the aminoalcohol lipidoid compound. These primary or secondary amines are left as is or may be reacted with another electrophile such as a different epoxide-terminated compound. As will be appreciated by one skilled in the art, reacting an amine with less than excess of epoxide-terminated compound will result in a plurality of different aminoalcohol lipidoid compounds with various numbers of tails. Certain amines may be fully functionalized with two epoxide-derived compound tails while other molecules will not be completely functionalized with epoxide-derived compound tails. For example, a diamine or polyamine may include one, two, three, or four epoxide-derived compound tails off the various amino moieties of the molecule resulting in primary, secondary, and tertiary amines. In certain embodiments, all the amino groups are not fully functionalized. In certain embodiments, two of the same types of epoxide-terminated compounds are used. In other embodiments, two or more different epoxide-terminated compounds are used. The synthesis of the aminoalcohol lipidoid compounds is performed with or without solvent, and the synthesis may be performed at higher temperatures ranging from 30-100 °C., preferably at approximately 50-90 °C. The prepared aminoalcohol lipidoid compounds may be optionally purified. For example, the mixture of aminoalcohol lipidoid compounds may be purified to yield an aminoalcohol lipidoid compound with a particular number of epoxide-derived compound tails. Or the mixture may be purified to yield a particular stereo- or regioisomer. The

aminoalcohol lipidoid compounds may also be alkylated using an alkyl halide (e.g., methyl iodide) or other alkylating agent, and/or they may be acylated.

[00443] US Patent Publication No. 20110293703 also provides libraries of aminoalcohol lipidoid compounds prepared by the inventive methods. These aminoalcohol lipidoid compounds may be prepared and/or screened using high-throughput techniques involving liquid handlers, robots, microtiter plates, computers, etc. In certain embodiments, the aminoalcohol lipidoid compounds are screened for their ability to transfect polynucleotides or other agents (e.g., proteins, peptides, small molecules) into the cell.

[00444] US Patent Publication No. 20130302401 relates to a class of poly(beta-amino alcohols) (PBAAs) has been prepared using combinatorial polymerization. The inventive PBAAs may be used in biotechnology and biomedical applications as coatings (such as coatings of films or multilayer films for medical devices or implants), additives, materials, excipients, non-biofouling agents, micropatterning agents, and cellular encapsulation agents. When used as surface coatings, these PBAAs elicited different levels of inflammation, both in vitro and in vivo, depending on their chemical structures. The large chemical diversity of this class of materials allowed us to identify polymer coatings that inhibit macrophage activation in vitro. Furthermore, these coatings reduce the recruitment of inflammatory cells, and reduce fibrosis, following the subcutaneous implantation of carboxylated polystyrene microparticles. These polymers may be used to form polyelectrolyte complex capsules for cell encapsulation. The invention may also have many other biological applications such as antimicrobial coatings, DNA or siRNA delivery, and stem cell tissue engineering. The teachings of US Patent Publication No. 20130302401 may be applied to the AD-functionalized CRISPR-Cas system of the present invention.

[00445] Preassembled recombinant CRISPR-Cas complexes comprising Cpf1, adenosine deaminase (which may be fused to Cpf1 or an adaptor protein), and guide RNA may be transfected, for example by electroporation, resulting in high mutation rates and absence of detectable off-target mutations. Hur, J.K. et al, Targeted mutagenesis in mice by electroporation of Cpf1 ribonucleoproteins, Nat Biotechnol. 2016 Jun 6. doi: 10.1038/nbt.3596.

[00446] In terms of local delivery to the brain, this can be achieved in various ways. For instance, material can be delivered intrastriatally e.g. by injection. Injection can be performed stereotactically via a craniotomy.

[00447] In some embodiments, sugar-based particles may be used, for example GalNAc, as described herein and with reference to WO2014118272 (incorporated herein by

reference) and Nair, JK et al., 2014, Journal of the American Chemical Society 136 (49), 16958-16961) and the teaching herein, especially in respect of delivery applies to all particles unless otherwise apparent. This may be considered to be a sugar-based particle and further details on other particle delivery systems and/or formulations are provided herein. GalNAc can therefore be considered to be a particle in the sense of the other particles described herein, such that general uses and other considerations, for instance delivery of said particles, apply to GalNAc particles as well. A solution-phase conjugation strategy may for example be used to attach triantennary GalNAc clusters (mol. wt. ~2000) activated as PFP (pentafluorophenyl) esters onto 5′-hexylamino modified oligonucleotides (5′-HA ASOs, mol. wt. ~8000 Da; Østergaard et al., Bioconjugate Chem., 2015, 26 (8), pp 1451–1455). Similarly, poly(acrylate) polymers have been described for in vivo nucleic acid delivery (see WO2013158141 incorporated herein by reference). In further alternative embodiments, pre-mixing CRISPR nanoparticles (or protein complexes) with naturally occurring serum proteins may be used in order to improve delivery (Akinc A et al, 2010, Molecular Therapy vol. 18 no. 7, 1357–1364).

[00448] Nanoclews

[00449] Further, the AD-functionalized CRISPR system may be delivered using nanoclews, for example as described in Sun W et al, *Cocoon-like self-degradable DNA nanoclew for anticancer drug delivery.,* J Am Chem Soc. 2014 Oct 22;136(42):14722-5. doi: 10.1021/ja5088024. Epub 2014 Oct 13. ; or in Sun W et al, *Self-Assembled DNA Nanoclews for the Efficient Delivery of CRISPR-Cas9 for Genome Editing.,* Angew Chem Int Ed Engl. 2015 Oct 5;54(41):12029-33. doi: 10.1002/anie.201506030. Epub 2015 Aug 27.

[00450] LNP

[00451] In some embodiments, delivery is by encapsulation of the Cpf1 protein or mRNA form in a lipid particle such as an LNP. In some embodiments, therefore, lipid nanoparticles (LNPs) are contemplated. An antitransthyretin small interfering RNA has been encapsulated in lipid nanoparticles and delivered to humans (see, e.g., Coelho et al., N Engl J Med 2013;369:819-29), and such a system may be adapted and applied to the CRISPR Cas system of the present invention. Doses of about 0.01 to about 1 mg per kg of body weight administered intravenously are contemplated. Medications to reduce the risk of infusion-related reactions are contemplated, such as dexamethasone, acetampinophen, diphenhydramine or cetirizine, and ranitidine are contemplated. Multiple doses of about 0.3 mg per kilogram every 4 weeks for five doses are also contemplated.

[00452] LNPs have been shown to be highly effective in delivering siRNAs to the liver (see, e.g., Tabernero et al., Cancer Discovery, April 2013, Vol. 3, No. 4, pages 363-470) and are therefore contemplated for delivering RNA encoding CRISPR Cas to the liver. A dosage of about four doses of 6 mg/kg of the LNP every two weeks may be contemplated. Tabernero et al. demonstrated that tumor regression was observed after the first 2 cycles of LNPs dosed at 0.7 mg/kg, and by the end of 6 cycles the patient had achieved a partial response with complete regression of the lymph node metastasis and substantial shrinkage of the liver tumors. A complete response was obtained after 40 doses in this patient, who has remained in remission and completed treatment after receiving doses over 26 months. Two patients with RCC and extrahepatic sites of disease including kidney, lung, and lymph nodes that were progressing following prior therapy with VEGF pathway inhibitors had stable disease at all sites for approximately 8 to 12 months, and a patient with PNET and liver metastases continued on the extension study for 18 months (36 doses) with stable disease.

[00453] However, the charge of the LNP must be taken into consideration. As cationic lipids combined with negatively charged lipids to induce nonbilayer structures that facilitate intracellular delivery. Because charged LNPs are rapidly cleared from circulation following intravenous injection, ionizable cationic lipids with pKa values below 7 were developed (see, e.g., Rosin et al, Molecular Therapy, vol. 19, no. 12, pages 1286-2200, Dec. 2011). Negatively charged polymers such as RNA may be loaded into LNPs at low pH values (e.g., pH 4) where the ionizable lipids display a positive charge. However, at physiological pH values, the LNPs exhibit a low surface charge compatible with longer circulation times. Four species of ionizable cationic lipids have been focused upon, namely 1,2-dilineoyl-3-dimethylammonium-propane (DLinDAP), 1,2-dilinoleyloxy-3-N,N-dimethylaminopropane (DLinDMA), 1,2-dilinoleyloxy-keto-N,N-dimethyl-3-aminopropane (DLinKDMA), and 1,2-dilinoleyl-4-(2-dimethylaminoethyl)-[1,3]-dioxolane (DLinKC2-DMA). It has been shown that LNP siRNA systems containing these lipids exhibit remarkably different gene silencing properties in hepatocytes in vivo, with potencies varying according to the series DLinKC2-DMA>DLinKDMA>DLinDMA>>DLinDAP employing a Factor VII gene silencing model (see, e.g., Rosin et al, Molecular Therapy, vol. 19, no. 12, pages 1286-2200, Dec. 2011). A dosage of 1 μg/ml of LNP or CRISPR-Cas RNA in or associated with the LNP may be contemplated, especially for a formulation containing DLinKC2-DMA.

[00454] Preparation of LNPs and CRISPR Cas encapsulation may be used/and or adapted from Rosin et al, Molecular Therapy, vol. 19, no. 12, pages 1286-2200, Dec. 2011).

The cationic lipids 1,2-dilineoyl-3-dimethylammonium-propane (DLinDAP), 1,2-dilinoleyloxy-3-N,N-dimethylaminopropane (DLinDMA), 1,2-dilinoleyloxyketo-N,N-dimethyl-3-aminopropane (DLinK-DMA), 1,2-dilinoleyl-4-(2-dimethylaminoethyl)-[1,3]-dioxolane (DLinKC2-DMA), (3-o-[2″-(methoxypolyethyleneglycol 2000) succinoyl]-1,2-dimyristoyl-sn-glycol (PEG-S-DMG), and R-3-[(ω-methoxy-poly(ethylene glycol)2000) carbamoyl]-1,2-dimyristyloxlpropyl-3-amine (PEG-C-DOMG) may be provided by Tekmira Pharmaceuticals (Vancouver, Canada) or synthesized. Cholesterol may be purchased from Sigma (St Louis, MO). The specific CRISPR Cas RNA may be encapsulated in LNPs containing DLinDAP, DLinDMA, DLinK-DMA, and DLinKC2-DMA (cationic lipid:DSPC:CHOL: PEGS-DMG or PEG-C-DOMG at 40:10:40:10 molar ratios). When required, 0.2% SP-DiOC18 (Invitrogen, Burlington, Canada) may be incorporated to assess cellular uptake, intracellular delivery, and biodistribution. Encapsulation may be performed by dissolving lipid mixtures comprised of cationic lipid:DSPC:cholesterol:PEG-c-DOMG (40:10:40:10 molar ratio) in ethanol to a final lipid concentration of 10 mmol/l. This ethanol solution of lipid may be added drop-wise to 50 mmol/l citrate, pH 4.0 to form multilamellar vesicles to produce a final concentration of 30% ethanol vol/vol. Large unilamellar vesicles may be formed following extrusion of multilamellar vesicles through two stacked 80 nm Nuclepore polycarbonate filters using the Extruder (Northern Lipids, Vancouver, Canada). Encapsulation may be achieved by adding RNA dissolved at 2 mg/ml in 50 mmol/l citrate, pH 4.0 containing 30% ethanol vol/vol drop-wise to extruded preformed large unilamellar vesicles and incubation at 31 °C for 30 minutes with constant mixing to a final RNA/lipid weight ratio of 0.06/1 wt/wt. Removal of ethanol and neutralization of formulation buffer were performed by dialysis against phosphate-buffered saline (PBS), pH 7.4 for 16 hours using Spectra/Por 2 regenerated cellulose dialysis membranes. Nanoparticle size distribution may be determined by dynamic light scattering using a NICOMP 370 particle sizer, the vesicle/intensity modes, and Gaussian fitting (Nicomp Particle Sizing, Santa Barbara, CA). The particle size for all three LNP systems may be ~70 nm in diameter. RNA encapsulation efficiency may be determined by removal of free RNA using VivaPureD MiniH columns (Sartorius Stedim Biotech) from samples collected before and after dialysis. The encapsulated RNA may be extracted from the eluted nanoparticles and quantified at 260 nm. RNA to lipid ratio was determined by measurement of cholesterol content in vesicles using the Cholesterol E enzymatic assay from Wako Chemicals USA (Richmond, VA). In conjunction with the

herein discussion of LNPs and PEG lipids, PEGylated liposomes or LNPs are likewise suitable for delivery of a CRISPR-Cas system or components thereof.

[00455] A lipid premix solution (20.4 mg/ml total lipid concentration) may be prepared in ethanol containing DLinKC2-DMA, DSPC, and cholesterol at 50:10:38.5 molar ratios. Sodium acetate may be added to the lipid premix at a molar ratio of 0.75:1 (sodium acetate:DLinKC2-DMA). The lipids may be subsequently hydrated by combining the mixture with 1.85 volumes of citrate buffer (10 mmol/l, pH 3.0) with vigorous stirring, resulting in spontaneous liposome formation in aqueous buffer containing 35% ethanol. The liposome solution may be incubated at 37 °C to allow for time-dependent increase in particle size. Aliquots may be removed at various times during incubation to investigate changes in liposome size by dynamic light scattering (Zetasizer Nano ZS, Malvern Instruments, Worcestershire, UK). Once the desired particle size is achieved, an aqueous PEG lipid solution (stock = 10 mg/ml PEG-DMG in 35% (vol/vol) ethanol) may be added to the liposome mixture to yield a final PEG molar concentration of 3.5% of total lipid. Upon addition of PEG-lipids, the liposomes should their size, effectively quenching further growth. RNA may then be added to the empty liposomes at an RNA to total lipid ratio of approximately 1:10 (wt:wt), followed by incubation for 30 minutes at 37 °C to form loaded LNPs. The mixture may be subsequently dialyzed overnight in PBS and filtered with a 0.45-μm syringe filter.

[00456] Spherical Nucleic Acid (SNA™) constructs and other nanoparticles (particularly gold nanoparticles) are also contemplated as a means to delivery CRISPR-Cas system to intended targets. Significant data show that AuraSense Therapeutics' Spherical Nucleic Acid (SNA™) constructs, based upon nucleic acid-functionalized gold nanoparticles, are useful.

[00457] Literature that may be employed in conjunction with herein teachings include: Cutler et al., J. Am. Chem. Soc. 2011 133:9254-9257, Hao et al., Small. 2011 7:3158-3162, Zhang et al., ACS Nano. 2011 5:6962-6970, Cutler et al., J. Am. Chem. Soc. 2012 134:1376-1391, Young et al., Nano Lett. 2012 12:3867-71, Zheng et al., Proc. Natl. Acad. Sci. USA. 2012 109:11975-80, Mirkin, Nanomedicine 2012 7:635-638 Zhang et al., J. Am. Chem. Soc. 2012 134:16488-1691, Weintraub, Nature 2013 495:S14-S16, Choi et al., Proc. Natl. Acad. Sci. USA. 2013 110(19):7625-7630, Jensen et al., Sci. Transl. Med. 5, 209ra152 (2013) and Mirkin, et al., Small, 10:186-192.

[00458] Self-assembling nanoparticles with RNA may be constructed with polyethyleneimine (PEI) that is PEGylated with an Arg-Gly-Asp (RGD) peptide ligand

attached at the distal end of the polyethylene glycol (PEG). This system has been used, for example, as a means to target tumor neovasculature expressing integrins and deliver siRNA inhibiting vascular endothelial growth factor receptor-2 (VEGF R2) expression and thereby achieve tumor angiogenesis (see, e.g., Schiffelers et al., Nucleic Acids Research, 2004, Vol. 32, No. 19). Nanoplexes may be prepared by mixing equal volumes of aqueous solutions of cationic polymer and nucleic acid to give a net molar excess of ionizable nitrogen (polymer) to phosphate (nucleic acid) over the range of 2 to 6. The electrostatic interactions between cationic polymers and nucleic acid resulted in the formation of polyplexes with average particle size distribution of about 100 nm, hence referred to here as nanoplexes. A dosage of about 100 to 200 mg of CRISPR Cas is envisioned for delivery in the self-assembling nanoparticles of Schiffelers et al.

[00459] The nanoplexes of Bartlett et al. (PNAS, September 25, 2007,vol. 104, no. 39) may also be applied to the present invention. The nanoplexes of Bartlett et al. are prepared by mixing equal volumes of aqueous solutions of cationic polymer and nucleic acid to give a net molar excess of ionizable nitrogen (polymer) to phosphate (nucleic acid) over the range of 2 to 6. The electrostatic interactions between cationic polymers and nucleic acid resulted in the formation of polyplexes with average particle size distribution of about 100 nm, hence referred to here as nanoplexes. The DOTA-siRNA of Bartlett et al. was synthesized as follows: 1,4,7,10-tetraazacyclododecane-1,4,7,10-tetraacetic acid mono(N-hydroxysuccinimide ester) (DOTA-NHSester) was ordered from Macrocyclics (Dallas, TX). The amine modified RNA sense strand with a 100-fold molar excess of DOTA-NHS-ester in carbonate buffer (pH 9) was added to a microcentrifuge tube. The contents were reacted by stirring for 4 h at room temperature. The DOTA-RNAsense conjugate was ethanol-precipitated, resuspended in water, and annealed to the unmodified antisense strand to yield DOTA-siRNA. All liquids were pretreated with Chelex-100 (Bio-Rad, Hercules, CA) to remove trace metal contaminants. Tf-targeted and nontargeted siRNA nanoparticles may be formed by using cyclodextrin-containing polycations. Typically, nanoparticles were formed in water at a charge ratio of 3 (+/-) and an siRNA concentration of 0.5 g/liter. One percent of the adamantane-PEG molecules on the surface of the targeted nanoparticles were modified with Tf (adamantane-PEG-Tf). The nanoparticles were suspended in a 5% (wt/vol) glucose carrier solution for injection.

[00460] Davis et al. (Nature, Vol 464, 15 April 2010) conducts a RNA clinical trial that uses a targeted nanoparticle-delivery system (clinical trial registration number NCT00689065). Patients with solid cancers refractory to standard-of-care therapies are

administered doses of targeted nanoparticles on days 1, 3, 8 and 10 of a 21-day cycle by a 30-min intravenous infusion. The nanoparticles consist of a synthetic delivery system containing: (1) a linear, cyclodextrin-based polymer (CDP), (2) a human transferrin protein (TF) targeting ligand displayed on the exterior of the nanoparticle to engage TF receptors (TFR) on the surface of the cancer cells, (3) a hydrophilic polymer (polyethylene glycol (PEG) used to promote nanoparticle stability in biological fluids), and (4) siRNA designed to reduce the expression of the RRM2 (sequence used in the clinic was previously denoted siR2B+5). The TFR has long been known to be upregulated in malignant cells, and RRM2 is an established anti-cancer target. These nanoparticles (clinical version denoted as CALAA-01) have been shown to be well tolerated in multi-dosing studies in non-human primates. Although a single patient with chronic myeloid leukaemia has been administered siRNA by liposomal delivery, Davis et al.'s clinical trial is the initial human trial to systemically deliver siRNA with a targeted delivery system and to treat patients with solid cancer. To ascertain whether the targeted delivery system can provide effective delivery of functional siRNA to human tumors, Davis et al. investigated biopsies from three patients from three different dosing cohorts; patients A, B and C, all of whom had metastatic melanoma and received CALAA-01 doses of 18, 24 and 30 mg m^{-2} siRNA, respectively. Similar doses may also be contemplated for the CRISPR Cas system of the present invention. The delivery of the invention may be achieved with nanoparticles containing a linear, cyclodextrin-based polymer (CDP), a human transferrin protein (TF) targeting ligand displayed on the exterior of the nanoparticle to engage TF receptors (TFR) on the surface of the cancer cells and/or a hydrophilic polymer (for example, polyethylene glycol (PEG) used to promote nanoparticle stability in biological fluids).

[00461] US Patent No. 8,709,843, incorporated herein by reference, provides a drug delivery system for targeted delivery of therapeutic agent-containing particles to tissues, cells, and intracellular compartments. The invention provides targeted particles comprising comprising polymer conjugated to a surfactant, hydrophilic polymer or lipid. US Patent No. 6,007,845, incorporated herein by reference, provides particles which have a core of a multiblock copolymer formed by covalently linking a multifunctional compound with one or more hydrophobic polymers and one or more hydrophilic polymers, and conatin a biologically active material. US Patent No. 5,855,913, incorporated herein by reference, provides a particulate composition having aerodynamically light particles having a tap density of less than 0.4 g/cm3 with a mean diameter of between 5 μm and 30 μ m, incorporating a surfactant on the surface thereof for drug delivery to the pulmonary system.

US Patent No. 5,985,309, incorporated herein by reference, provides particles incorporating a surfactant and/or a hydrophilic or hydrophobic complex of a positively or negatively charged therapeutic or diagnostic agent and a charged molecule of opposite charge for delivery to the pulmonary system. US. Patent No. 5,543,158, incorporated herein by reference, provides biodegradable injectable particles having a biodegradable solid core containing a biologically active material and poly(alkylene glycol) moieties on the surface. WO2012135025 (also published as US20120251560), incorporated herein by reference, describes conjugated polyethyleneimine (PEI) polymers and conjugated aza-macrocycles (collectively referred to as "conjugated lipomer" or "lipomers"). In certain embodiments, it can envisioned that such conjugated lipomers can be used in the context of the CRISPR-Cas system to achieve in vitro, ex vivo and in vivo genomic perturbations to modify gene expression, including modulation of protein expression.

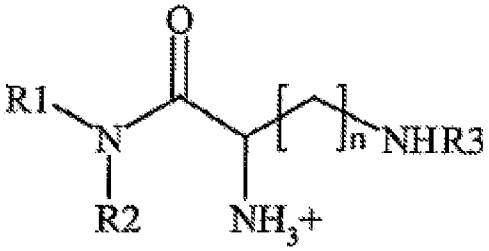
[00462] In one embodiment, the nanoparticle may be epoxide-modified lipid–polymer, advantageously 7C1 (see, e.g., James E. Dahlman and Carmen Barnes et al. Nature Nanotechnology (2014) published online 11 May 2014, doi:10.1038/nnano.2014.84). C71 was synthesized by reacting C15 epoxide-terminated lipids with PEI600 at a 14:1 molar ratio, and was formulated with C14PEG2000 to produce nanoparticles (diameter between 35 and 60 nm) that were stable in PBS solution for at least 40 days.

[00463] An epoxide-modified lipid-polymer may be utilized to deliver the CRISPR-Cas system of the present invention to pulmonary, cardiovascular or renal cells, however, one of skill in the art may adapt the system to deliver to other target organs. Dosage ranging from about 0.05 to about 0.6 mg/kg are envisioned. Dosages over several days or weeks are also envisioned, with a total dosage of about 2 mg/kg.

[00464] In some embodiments, the LNP for deliverting the RNA molecules is prepared by methods known in the art, such as those described in, for example, WO 2005/105152 (PCT/EP2005/004920), WO 2006/069782 (PCT/EP2005/014074), WO 2007/121947 (PCT/EP2007/003496), and WO 2015/082080 (PCT/EP2014/003274), which are herein incorporated by reference. LNPs aimed specifically at the enhanced and improved delivery of siRNA into mammalian cells are described in, for example, Aleku *et al.*, *Cancer Res.*, 68(23): 9788-98 (Dec. 1, 2008), Strumberg *et al.*, *Int. J. Clin. Pharmacol. Ther.*, 50(1): 76-8 (Jan. 2012), Schultheis *et al.*, *J. Clin. Oncol.*, 32(36): 4141-48 (Dec. 20, 2014), and Fehring *et al.*, *Mol. Ther.*, 22(4): 811-20 (Apr. 22, 2014), which are herein incorporated by reference and may be applied to the present technology.

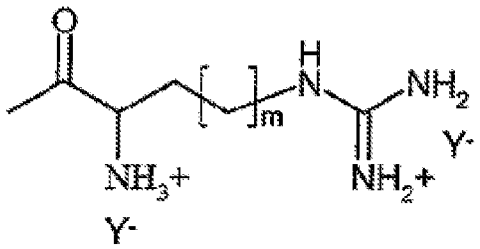
[00465] In some embodiments, the LNP includes any LNP disclosed in WO 2005/105152 (PCT/EP2005/004920), WO 2006/069782 (PCT/EP2005/014074), WO 2007/121947 (PCT/EP2007/003496), and WO 2015/082080 (PCT/EP2014/003274).

[00466] In some embodiments, the LNP includes at least one lipid having Formula I:



(Formula I),

wherein R1 and R2 are each and independently selected from the group comprising alkyl, n is any integer between 1 and 4, and R3 is an acyl selected from the group comprising lysyl, ornithyl, 2,4-diaminobutyryl, histidyl and an acyl moiety according to Formula II:

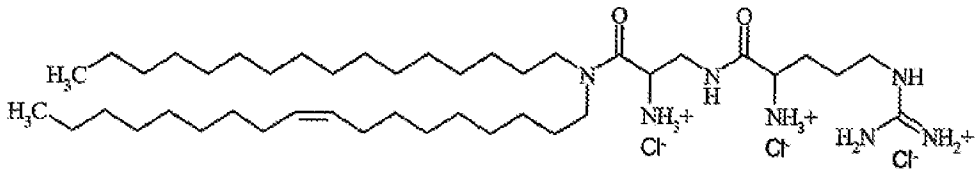


(Formula II),

wherein m is any integer from 1 to 3 and Y^- is a pharmaceutically acceptable anion. In some embodiments, a lipid according to Formula I includes at least two asymmetric C atoms. In some embodiments, enantiomers of Formula I include, but are not limited to, R-R; S-S; R-S and S-R enantiomer.

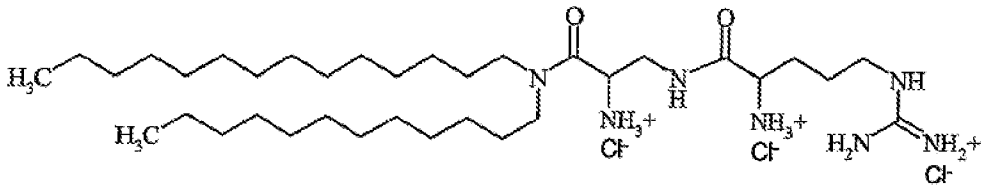
[00467] In some embodiments, R1 is lauryl and R2 is myristyl. In another embodiment, R1 is palmityl and R2 is oleyl. In some embodiments, m is 1 or 2. In some embodiments, Y^- is selected from halogenids, acetate or trifluoroacetate.

[00468] In some embodiments, the LNP comprises one or more lipids select from: □-arginyl-2,3-diamino propionic acid-N-palmityl-N-oleyl-amide trihydrochloride (Formula III):



(Formula III);

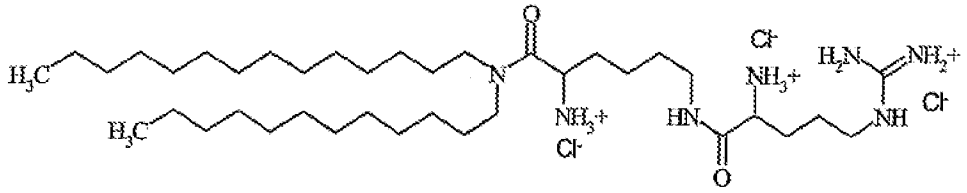
□-arginyl-2,3-diamino propionic acid-N-lauryl-N-myristyl-amide trihydrochloride (Formula IV):



(Formula IV);

and

□-arginyl-lysine-N-lauryl-N-myristyl-amide trihydrochloride (Formula V):



(Formula V).

[00469] In some embodiments, the LNP also includes a constituent. By way of example, but not by way of limitation, in some embodiments, the constituent is selected from peptides, proteins, oligonucleotides, polynucleotides, nucleic acids, or a combination thereof. In some embodiments, the constituent is an antibody, *e.g.*, a monoclonal antibody. In some embodiments, the constituent is a nucleic acid selected from, *e.g.*, ribozymes, aptamers, spiegelmers, DNA, RNA, PNA, LNA, or a combination thereof. In some embodiments, the nucleic acid is guide RNA and/or mRNA.

[00470] In some embodiments, the constituent of the LNP comprises an mRNA encoding a CRIPSR-Cas protein. In some embodiments, the constituent of the LNP comprises an mRNA encoding a Type-II or Type-V CRIPSR-Cas protein. In some embodiments, the constituent of the LNP comprises an mRNA encoding an adenosine deaminase (which may be fused to a CRISPR-Cas protein or an adaptor protein).

[00471] In some embodiments, the constituent of the LNP further comprises one or more guide RNA. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to vascular endothelium. In some embodiments, the LNP is

configured to deliver the aforementioned mRNA and guide RNA to pulmonary endothelium. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to liver. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to lung. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to hearts. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to spleen. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to kidney. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to pancrea. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to brain. In some embodiments, the LNP is configured to deliver the aforementioned mRNA and guide RNA to macrophages.

[00472] In some embodiments, the LNP also includes at least one helper lipid. In some embodiments, the helper lipid is selected from phospholipids and steroids. In some embodiments, the phospholipids are di- and /or monoester of the phosphoric acid. In some embodiments, the phospholipids are phosphoglycerides and /or sphingolipids. In some embodiments, the steroids are naturally occurring and/or synthetic compounds based on the partially hydrogenated cyclopenta[a]phenanthrene. In some embodiments, the steroids contain 21 to 30 C atoms. In some embodiments, the steroid is cholesterol. In some embodiments, the helper lipid is selected from 1,2-diphytanoyl-sn-glycero-3-phosphoethanolamine (DPhyPE), ceramide, and 1,2-dioleylsn-glycero-3-phosphoethanolamine (DOPE).

[00473] In some embodiments, the at least one helper lipid comprises a moiety selected from the group comprising a PEG moiety, a HEG moiety, a polyhydroxyethyl starch (polyHES) moiety and a polypropylene moiety. In some embodiments, the moiety has a molecule weight between about 500 to 10,000 Da or between about 2,000 to 5,000 Da. In some embodiments, the PEG moiety is selected from 1,2-distearoyl-sn-glycero-3 phosphoethanolamine, 1,2-dialkyl-sn-glycero-3-phosphoethanolamine, and Ceramide-PEG. In some embodiments, the PEG moiety has a molecular weight between about 500 to 10,000 Da or between about 2,000 to 5,000 Da. In some embodiments, the PEG moiety has a molecular weight of 2,000 Da.

[00474] In some embodiments, the helper lipid is between about 20 mol % to 80 mol % of the total lipid content of the composition. In some embodiments, the helper lipid component is between about 35 mol % to 65 mol % of the total lipid content of the LNP. In

some embodiments, the LNP includes lipids at 50 mol% and the helper lipid at 50 mol% of the total lipid content of the LNP.

[00475] In some embodiments, the LNP includes any of □-3-arginyl-2,3-diaminopropionic acid-N-palmityl-N-oleyl-amide trihydrochloride, □-arginyl-2,3-diaminopropionic acid-N-lauryl-N-myristyl-amide trihydrochloride or □-arginyl-lysine-N-lauryl-N-myristyl-amide trihydrochloride in combination with DPhyPE, wherein the content of DPhyPE is about 80 mol %, 65 mol %, 50 mol % and 35 mol % of the overall lipid content of the LNP. In some embodiments, the LNP includes □-arginyl-2,3-diamino propionic acid-N-pahnityl-N-oleyl-amide trihydrochloride (lipid) and 1,2-diphytanoyl-sn-glycero-3-phosphoethanolamine (helper lipid). In some embodiments, the LNP includes □-arginyl-2,3-diamino propionic acid-N-palmityl-N-oleyl-amide trihydrochloride (lipid), l,2-diphytanoyl-sn-glycero-3-phosphoethanolamine (first helper lipid), and l,2-disteroyl-sn-glycero-3-phosphoethanolamine-PEG2000 (second helper lipid).

[00476] In some embodiments, the second helper lipid is between about 0.05 mol% to 4.9 mol% or between about 1 mol% to 3 mol% of the total lipid content. In some embodiments, the LNP includes lipids at between about 45 mol% to 50 mol% of the total lipid content, a first helper lipid between about 45 mol% to 50 mol% of the total lipid content, under the proviso that there is a PEGylated second helper lipid between about 0.1 mol% to 5 mol %, between about 1 mol% to 4 mol%, or at about 2 mol% of the total lipid content, wherein the sum of the content of the lipids, the first helper lipid, and of the second helper lipid is 100 mol% of the total lipid content and wherein the sum of the first helper lipid and the second helper lipid is 50 mol% of the total lipid content. In some embodiments, the LNP comprises: (a) 50 mol% of □-arginyl-2,3-diamino propionic acid-N-palmityl-N-oleyl-amide trihydrochloride, 48 mol% of l,2-diphytanoyl-sn-glycero-3-phosphoethanolamine; and 2 mol% l,2-distearoyl-sn-glycero-3-phosphoethanolamine-PEG2000; or (b) 50 mol% of □-arginyl-2,3-diamino propionic acid-N-palmityl-N-oleyl-amide trihydrocloride, 49 mol% l,2-diphytanoyl-sn-glycero-3-phosphoethanolamine; and 1 mol% N(Carbonyl-methoxypolyethylenglycol-2000)-1,2-distearoyl-sn-glycero3-phosphoethanolamine, or a sodium salt thereof.

[00477] In some embodiments, the LNP contains a nucleic acid, wherein the charge ratio of nucleic acid backbone phosphates to cationic lipid nitrogen atoms is about 1: 1.5 – 7 or about 1:4.

[00478] In some embodiments, the LNP also includes a shielding compound, which is removable from the lipid composition under *in vivo* conditions. In some embodiments, the

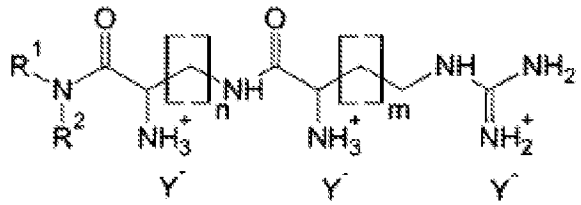
shielding compound is a biologically inert compound. In some embodiments, the shielding compound does not carry any charge on its surface or on the molecule as such. In some embodiments, the shielding compounds are polyethylenglycoles (PEGs), hydroxyethylglucose (HEG) based polymers, polyhydroxyethyl starch (polyHES) and polypropylene. In some embodiments, the PEG, HEG, polyHES, and a polypropylene weight between about 500 to 10,000 Da or between about 2000 to 5000 Da. In some embodiments, the shielding compound is PEG2000 or PEG5000.

[00479] In some embodiments, the LNP includes at least one lipid, a first helper lipid, and a shielding compound that is removable from the lipid composition under *in vivo* conditions. In some embodiments, the LNP also includes a second helper lipid. In some embodiments, the first helper lipid is ceramide. In some embodiments, the second helper lipid is ceramide. In some embodiments, the ceramide comprises at least one short carbon chain substituent of from 6 to 10 carbon atoms. In some embodiments, the ceramide comprises 8 carbon atoms. In some embodiments, the shielding compound is attached to a ceramide. In some embodiments, the shielding compound is attached to a ceramide. In some embodiments, the shielding compound is covalently attached to the ceramide. In some embodiments, the shielding compound is attached to a nucleic acid in the LNP. In some embodiments, the shielding compound is covalently attached to the nucleic acid. In some embodiments, the shielding compound is attached to the nucleic acid by a linker. In some embodiments, the linker is cleaved under physiological conditions. In some embodiments, the linker is selected from ssRNA, ssDNA, dsRNA, dsDNA, peptide, S-S-linkers and pH sensitive linkers. In some embodiments, the linker moiety is attached to the 3' end of the sense strand of the nucleic acid. In some embodiments, the shielding compound comprises a pH-sensitive linker or a pH-sensitive moiety. In some embodiments, the pH-sensitive linker or pH-sensitive moiety is an anionic linker or an anionic moiety. In some embodiments, the anionic linker or anionic moiety is less anionic or neutral in an acidic environment. In some embodiments, the pH-sensitive linker or the pH-sensitive moiety is selected from the oligo (glutamic acid), oligophenolate(s) and diethylene triamine penta acetic acid.

[00480] In any of the LNP embodiments in the previous paragraph, the LNP can have an osmolality between about 50 to 600 mosmole/kg, between about 250 to 350 mosmole/kg, or between about 280 to 320 mosmole/kg, and/or wherein the LNP formed by the lipid and/or one or two helper lipids and the shielding compound have a particle size between about 20 to 200 nm, between about 30 to 100 nm, or between about 40 to 80 nm.

[00481] In some embodiments, the shielding compound provides for a longer circulation time *in vivo* and allows for a better biodistribution of the nucleic acid containing LNP. In some embodiments, the shielding compound prevents immediate interaction of the LNP with serum compounds or compounds of other bodily fluids or cytoplasma membranes, *e.g.*, cytoplasma membranes of the endothelial lining of the vasculature, into which the LNP is administered. Additionally or alternatively, in some embodiments, the shielding compounds also prevent elements of the immune system from immediately interacting with the LNP. Additionally or alternatively, in some embodiments, the shielding compound acts as an anti-opsonizing compound. Without wishing to be bound by any mechanism or theory, in some embodiments, the shielding compound forms a cover or coat that reduces the surface area of the LNP available for interaction with its environment. Additionally or alternatively, in some embodiments, the shielding compound shields the overall charge of the LNP.

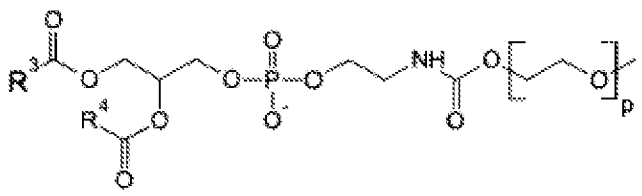
[00482] In another embodiment, the LNP includes at least one cationic lipid having Formula VI:



(Formula VI),

wherein n is 1, 2, 3, or 4, wherein m is 1, 2, or 3, wherein Y^- is anion, wherein each of R^1 and R^2 is individually and independently selected from the group consisting of linear C12-C18 alkyl and linear C12-C18 alkenyl, a sterol compound, wherein the sterol compound is selected from the group consisting of cholesterol and stigmasterol, and a PEGylated lipid, wherein the PEGylated lipid comprises a PEG moiety, wherein the PEGylated lipid is selected from the group consisting of:

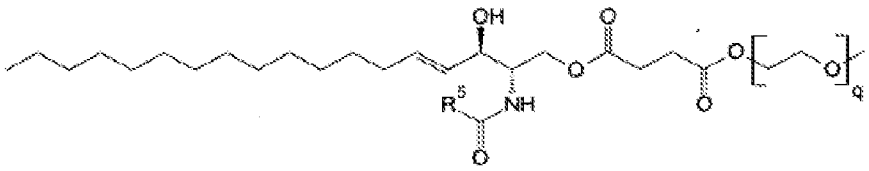
a PEGylated phosphoethanolamine of Formula VII:



(Formula VII),

wherein R^3 and R^4 are individually and independently linear C13-C17 alkyl, and p is any integer between 15 to 130;

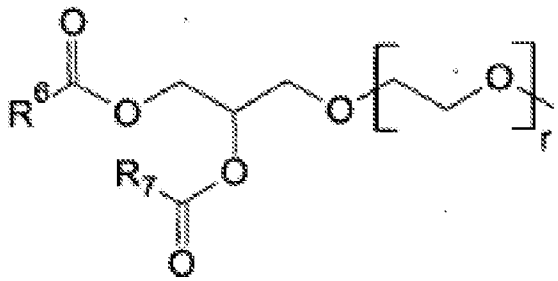
a PEGylated ceramide of Formula VIII:



(Formula VIII);

wherein R^5 is linear C7-C15 alkyl, and q is any number between 15 to 130; and

a PEGylated diacylglycerol of Formula IX:

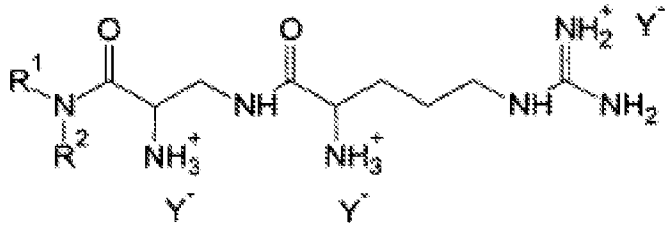


(Formula IX),

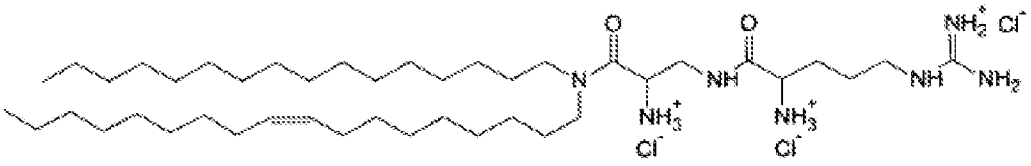
wherein each of R^6 and R^7 is individually and independently linear C11-C17 alkyl, and r is any integer from 15 to 130.

[00483] In some embodiments, R^1 and R^2 are different from each other. In some embodiments, R^1 is palmityl and R^2 is oleyl. In some embodiments, R^1 is lauryl and R^2 is myristyl. In some embodiments, R^1 and R^2 are the same. In some embodiments, each of R^1 and R^2 is individually and independently selected from the group consisting of C12 alkyl, C14 alkyl, Cl6 alkyl, Cl8 alkyl, Cl2 alkenyl, C14 alkenyl, Cl6 alkenyl and Cl8 alkenyl. In some embodiments, each of C12 alkenyl, C14 alkenyl, Cl6 alkenyl and Cl 8 alkenyl comprises one or two double bonds. In some embodiments, Cl8 alkenyl is Cl8 alkenyl with one double bond between C9 and C10. In some embodiments, C18 alkenyl is cis-9-octadecyl.

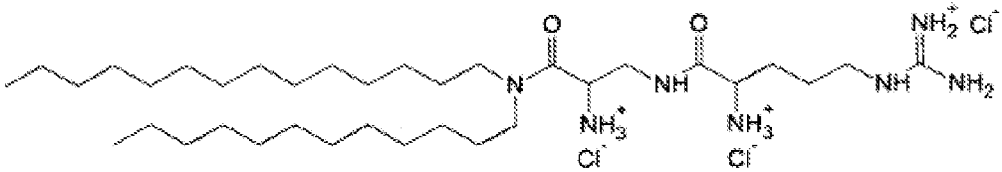
[00484] In some embodiments, the cationic lipid is a compound of Formula X:



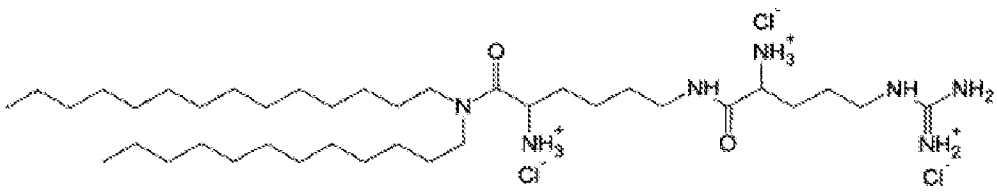
(Formula X). In some embodiments, Y^- is selected from halogenids, acetate and trifluoroacetate. In some embodiments, the cationic lipid is □-arginyl-2,3-diamino propionic acid-N-palmityl-N-oleyl-amide trihydrochloride of Formula III:



(Formula III). In some embodiments, the cationic lipid is □-arginyl-2,3-diamino propionic acid-N-lauryl-N-myristyl-amide trihydrochloride of Formula IV:



(Formula IV). In some embodiments, the cationic lipid is □-arginyl-lysine-N-lauryl-N-myristyl-amide trihydrochloride of Formula V:

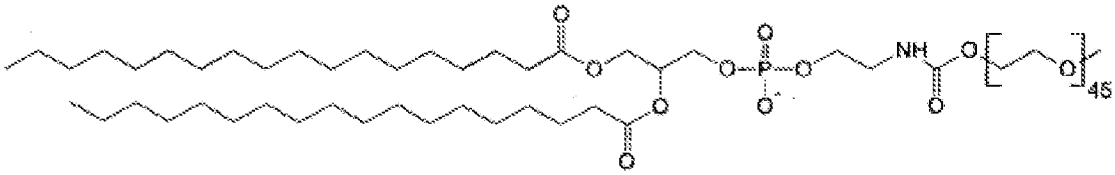


(Formula V).

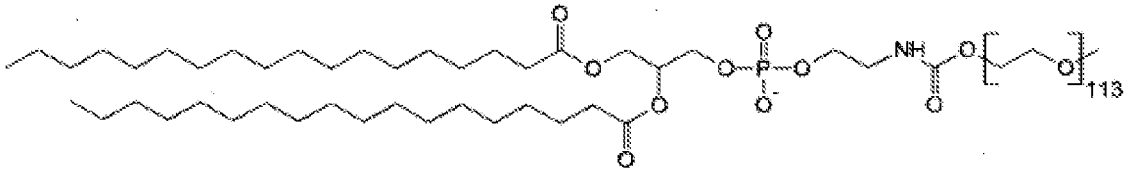
[00485] In some embodiments, the sterol compound is cholesterol. In some embodiments, the sterol compound is stigmasterin.

[00486] In some embodiments, the PEG moiety of the PEGylated lipid has a molecular weight from about 800 to 5,000 Da. In some embodiments, the molecular weight of the PEG moiety of the PEGylated lipid is about 800 Da. In some embodiments, the molecular weight of the PEG moiety of the PEGylated lipid is about 2,000 Da. In some embodiments, the molecular weight of the PEG moiety of the PEGylated lipid is about 5,000 Da.In some

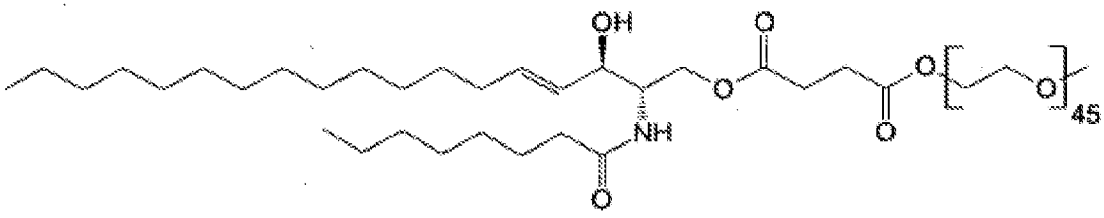
embodiments, the PEGylated lipid is a PEGylated phosphoethanolamine of Formula VII, wherein each of R^3 and R^4 is individually and independently linear C13-C17 alkyl, and p is any integer from 18, 19 or 20, or from 44, 45 or 46 or from 113, 114 or 115. In some embodiments, R^3 and R^4 are the same. In some embodiments, R^3 and R^4 are different. In some embodiments, each of R^3 and R^4 is individually and independently selected from the group consisting of C13 alkyl, Cl5 alkyl and Cl7 alkyl. In some embodiments, the PEGylated phosphoethanolamine of Formula VII is 1,2-distearoyl-*sn*-glycero-3-phosphoethanolamine-N-[methoxy(polyethylene glycol)-2000] (ammonium salt):



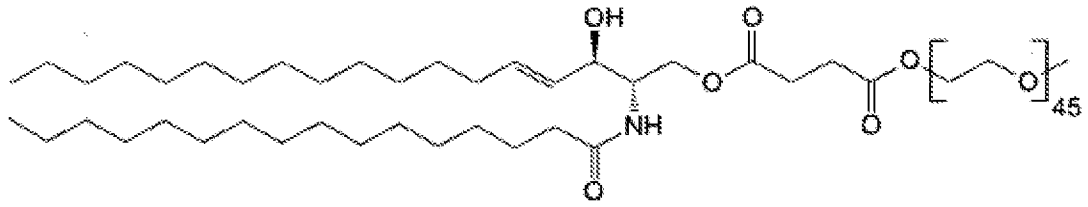
(Formula XI). In some embodiments, the PEGylated phosphoethanolamine of Formula VII is 1,2-distearoyl-*sn*-glycero-3-phosphoethanolamine-N-[methoxy(polyethylene glycol)-5000] (ammonium salt):



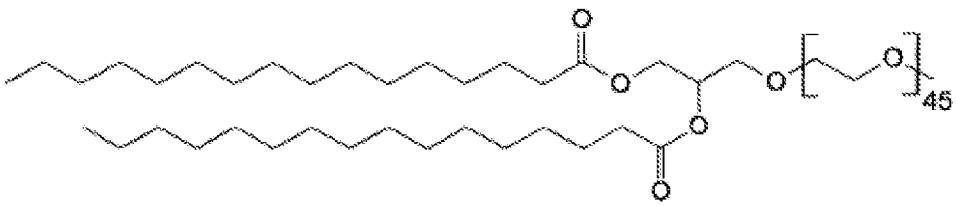
(Formula XII). In some embodiments, the PEGylated lipid is a PEGylated ceramide of Formula VIII, wherein R^5 is linear C7-C15 alkyl, and q is any integer from 18, 19 or 20, or from 44, 45 or 46 or from 113, 114 or 115. In some embodiments, R^5 is linear C7 alkyl. In some embodiments, R^5 is linear Cl5 alkyl.In some embodiments, the PEGylated ceramide of Formula VIII is N-octanoyl-sphingosine-1- {succinyl[methoxy(polyethylene glycol)2000]}:



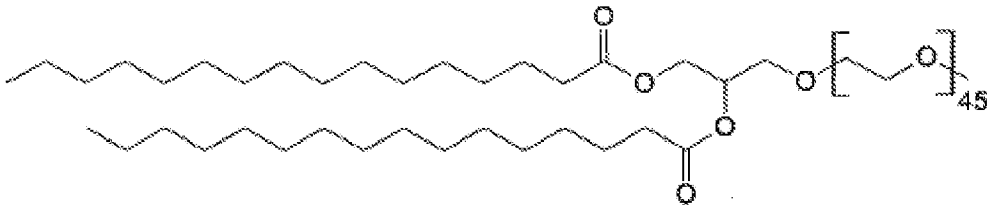
(Formula XIII). In some embodiments, the PEGylated ceramide of Formula VIII is N-palmitoyl-sphingosine-1- {succinyl[methoxy(polyethylene glycol)2000]}



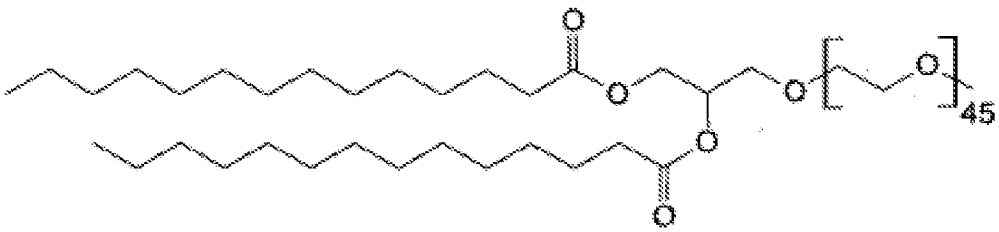
(Formula XIV). In some embodiments, the PEGylated lipid is a PEGylated diacylglycerol of Formula IX, wherein each of R^6 and R^7 is individually and independently linear C11-C17 alkyl, and r is any integer from 18, 19 or 20, or from 44, 45 or 46 or from 113, 114 or 115. In some embodiments, R^6 and R^7 are the same. In some embodiments, R^6 and R^7 are different. In some embodiments, each of R^6 and R^7 is individually and independently selected from the group consisting of linear C17 alkyl, linear C15 alkyl and linear C13 alkyl. In some embodiments, the PEGylated diacylglycerol of Formula IX 1,2-Distearoyl-sn-glycerol [methoxy(polyethylene glycol)2000]:



(Formula XV). In some embodiments, the PEGylated diacylglycerol of Formula IX is 1,2-Dipalmitoyl-sn-glycerol [methoxy(polyethylene glycol)2000]:



(Formula XVI). In some embodiments, the PEGylated diacylglycerol of Formula IX is:



(Formula XVII). In some embodiments, the LNP includes at least one cationic lipid selected from of

Formulas III, IV, and V, at least one sterol compound selected from a cholesterol and stigmasterin, and wherein the PEGylated lipid is at least one selected from Formulas XI and XII. In some embodiments, the LNP includes at least one cationic lipid selected from Formulas III, IV, and V, at least one sterol compound selected from a cholesterol and stigmasterin, and wherein the PEGylated lipid is at least one selected from Formulas XIII and XIV. In some embodiments, the LNP includes at least one cationic lipid selected from Formulas III, IV, and V, at least one sterol compound selected from a cholesterol and stigmasterin, and wherein the PEGylated lipid is at least one selected from Formulas XV and XVI. In some embodiments, the LNP includes a cationic lipid of Formula III, a cholesterol as the sterol compound, and wherein the PEGylated lipid is Formula XI.

[00487] In any of the LNP embodiments in the previous paragraph, wherein the content of the cationic lipid composition is between about 65 mole% to 75 mole%, the content of the sterol compound is between about 24 mole% to 34 mole% and the content of the PEGylated lipid is between about 0.5 mole% to 1.5 mole%, wherein the sum of the content of the cationic lipid, of the sterol compound and of the PEGylated lipid for the lipid composition is 100 mole%. In some embodiments, the cationic lipid is about 70 mole%, the content of the sterol compound is about 29 mole% and the content of the PEGylated lipid is about 1 mole%. In some embodiments, the LNP is 70 mole% of Formula III, 29 mole% of cholesterol, and 1 mole% of Formula XI.

[00488] Exosomes

[00489] Exosomes are endogenous nano-vesicles that transport RNAs and proteins, and which can deliver RNA to the brain and other target organs. To reduce immunogenicity, Alvarez-Erviti et al. (2011, Nat Biotechnol 29: 341) used self-derived dendritic cells for exosome production. Targeting to the brain was achieved by engineering the dendritic cells to express Lamp2b, an exosomal membrane protein, fused to the neuron-specific RVG peptide. Purified exosomes were loaded with exogenous RNA by electroporation. Intravenously injected RVG-targeted exosomes delivered GAPDH siRNA specifically to neurons, microglia, oligodendrocytes in the brain, resulting in a specific gene knockdown. Pre-exposure to RVG exosomes did not attenuate knockdown, and non-specific uptake in other tissues was not observed. The therapeutic potential of exosome-mediated siRNA delivery was demonstrated by the strong mRNA (60%) and protein (62%) knockdown of BACE1, a therapeutic target in Alzheimer's disease.

[00490] To obtain a pool of immunologically inert exosomes, Alvarez-Erviti et al. harvested bone marrow from inbred C57BL/6 mice with a homogenous major histocompatibility complex (MHC) haplotype. As immature dendritic cells produce large quantities of exosomes devoid of T-cell activators such as MHC-II and CD86, Alvarez-Erviti et al. selected for dendritic cells with granulocyte/macrophage-colony stimulating factor (GM-CSF) for 7 d. Exosomes were purified from the culture supernatant the following day using well-established ultracentrifugation protocols. The exosomes produced were physically homogenous, with a size distribution peaking at 80 nm in diameter as determined by nanoparticle tracking analysis (NTA) and electron microscopy. Alvarez-Erviti et al. obtained 6-12 μg of exosomes (measured based on protein concentration) per 10^6 cells.

[00491] Next, Alvarez-Erviti et al. investigated the possibility of loading modified exosomes with exogenous cargoes using electroporation protocols adapted for nanoscale applications. As electroporation for membrane particles at the nanometer scale is not well-characterized, nonspecific Cy5-labeled RNA was used for the empirical optimization of the electroporation protocol. The amount of encapsulated RNA was assayed after ultracentrifugation and lysis of exosomes. Electroporation at 400 V and 125 μF resulted in the greatest retention of RNA and was used for all subsequent experiments.

[00492] Alvarez-Erviti et al. administered 150 μg of each BACE1 siRNA encapsulated in 150 μg of RVG exosomes to normal C57BL/6 mice and compared the knockdown efficiency to four controls: untreated mice, mice injected with RVG exosomes only, mice injected with BACE1 siRNA complexed to an in vivo cationic liposome reagent and mice injected with BACE1 siRNA complexed to RVG-9R, the RVG peptide conjugated to 9 D-arginines that electrostatically binds to the siRNA. Cortical tissue samples were analyzed 3 d after administration and a significant protein knockdown (45%, $P < 0.05$, versus 62%, $P < 0.01$) in both siRNA-RVG-9R-treated and siRNARVG exosome-treated mice was observed, resulting from a significant decrease in BACE1 mRNA levels (66% [+ or -] 15%, $P < 0.001$ and 61% [+ or -] 13% respectively, $P < 0.01$). Moreover, Applicants demonstrated a significant decrease (55%, $P < 0.05$) in the total [beta]-amyloid 1-42 levels, a main component of the amyloid plaques in Alzheimer's pathology, in the RVG-exosome-treated animals. The decrease observed was greater than the β-amyloid 1-40 decrease demonstrated in normal mice after intraventricular injection of BACE1 inhibitors. Alvarez-Erviti et al. carried out 5'-rapid amplification of cDNA ends (RACE) on BACE1 cleavage product, which provided evidence of RNAi-mediated knockdown by the siRNA.

[00493] Finally, Alvarez-Erviti et al. investigated whether RNA-RVG exosomes induced immune responses *in vivo* by assessing IL-6, IP-10, TNFα and IFN-α serum concentrations. Following exosome treatment, nonsignificant changes in all cytokines were registered similar to siRNA-transfection reagent treatment in contrast to siRNA-RVG-9R, which potently stimulated IL-6 secretion, confirming the immunologically inert profile of the exosome treatment. Given that exosomes encapsulate only 20% of siRNA, delivery with RVG-exosome appears to be more efficient than RVG-9R delivery as comparable mRNA knockdown and greater protein knockdown was achieved with fivefold less siRNA without the corresponding level of immune stimulation. This experiment demonstrated the therapeutic potential of RVG-exosome technology, which is potentially suited for long-term silencing of genes related to neurodegenerative diseases. The exosome delivery system of Alvarez-Erviti et al. may be applied to deliver the AD-functionalized CRISPR-Cas system of the present invention to therapeutic targets, especially neurodegenerative diseases. A dosage of about 100 to 1000 mg of CRISPR Cas encapsulated in about 100 to 1000 mg of RVG exosomes may be contemplated for the present invention.

[00494] El-Andaloussi et al. (Nature Protocols 7,2112–2126(2012)) discloses how exosomes derived from cultured cells can be harnessed for delivery of RNA in vitro and in vivo. This protocol first describes the generation of targeted exosomes through transfection of an expression vector, comprising an exosomal protein fused with a peptide ligand. Next, El-Andaloussi et al. explain how to purify and characterize exosomes from transfected cell supernatant. Next, El-Andaloussi et al. detail crucial steps for loading RNA into exosomes. Finally, El-Andaloussi et al. outline how to use exosomes to efficiently deliver RNA in vitro and in vivo in mouse brain. Examples of anticipated results in which exosome-mediated RNA delivery is evaluated by functional assays and imaging are also provided. The entire protocol takes ~3 weeks. Delivery or administration according to the invention may be performed using exosomes produced from self-derived dendritic cells. From the herein teachings, this can be employed in the practice of the invention.

[00495] In another embodiment, the plasma exosomes of Wahlgren et al. (Nucleic Acids Research, 2012, Vol. 40, No. 17 e130) are contemplated. Exosomes are nano-sized vesicles (30–90nm in size) produced by many cell types, including dendritic cells (DC), B cells, T cells, mast cells, epithelial cells and tumor cells. These vesicles are formed by inward budding of late endosomes and are then released to the extracellular environment upon fusion with the plasma membrane. Because exosomes naturally carry RNA between cells,

this property may be useful in gene therapy, and from this disclosure can be employed in the practice of the instant invention.

[00496] Exosomes from plasma can be prepared by centrifugation of buffy coat at 900g for 20 min to isolate the plasma followed by harvesting cell supernatants, centrifuging at 300g for 10 min to eliminate cells and at 16 500g for 30 min followed by filtration through a 0.22 mm filter. Exosomes are pelleted by ultracentrifugation at 120 000g for70 min. Chemical transfection of siRNA into exosomes is carried out according to the manufacturer's instructions in RNAi Human/Mouse Starter Kit (Quiagen, Hilden, Germany). siRNA is added to 100 ml PBS at a final concentration of 2 mmol/ml. After adding HiPerFect transfection reagent, the mixture is incubated for 10 min at RT. In order to remove the excess of micelles, the exosomes are re-isolated using aldehyde/sulfate latex beads. The chemical transfection of CRISPR Cas into exosomes may be conducted similarly to siRNA. The exosomes may be co-cultured with monocytes and lymphocytes isolated from the peripheral blood of healthy donors. Therefore, it may be contemplated that exosomes containing CRISPR Cas may be introduced to monocytes and lymphocytes of and autologously reintroduced into a human. Accordingly, delivery or administration according to the invention may be performed using plasma exosomes.

[00497] Liposomes

[00498] Delivery or administration according to the invention can be performed with liposomes. Liposomes are spherical vesicle structures composed of a uni- or multilamellar lipid bilayer surrounding internal aqueous compartments and a relatively impermeable outer lipophilic phospholipid bilayer. Liposomes have gained considerable attention as drug delivery carriers because they are biocompatible, nontoxic, can deliver both hydrophilic and lipophilic drug molecules, protect their cargo from degradation by plasma enzymes, and transport their load across biological membranes and the blood brain barrier (BBB) (see, e.g., Spuch and Navarro, Journal of Drug Delivery, vol. 2011, Article ID 469679, 12 pages, 2011. doi:10.1155/2011/469679 for review).

[00499] Liposomes can be made from several different types of lipids; however, phospholipids are most commonly used to generate liposomes as drug carriers. Although liposome formation is spontaneous when a lipid film is mixed with an aqueous solution, it can also be expedited by applying force in the form of shaking by using a homogenizer, sonicator, or an extrusion apparatus (see, e.g., Spuch and Navarro, Journal of Drug Delivery, vol. 2011, Article ID 469679, 12 pages, 2011. doi:10.1155/2011/469679 for review).

[00500] Several other additives may be added to liposomes in order to modify their structure and properties. For instance, either cholesterol or sphingomyelin may be added to the liposomal mixture in order to help stabilize the liposomal structure and to prevent the leakage of the liposomal inner cargo. Further, liposomes are prepared from hydrogenated egg phosphatidylcholine or egg phosphatidylcholine, cholesterol, and dicetyl phosphate, and their mean vesicle sizes were adjusted to about 50 and 100 nm. (see, e.g., Spuch and Navarro, Journal of Drug Delivery, vol. 2011, Article ID 469679, 12 pages, 2011. doi:10.1155/2011/469679 for review).

[00501] A liposome formulation may be mainly comprised of natural phospholipids and lipids such as 1,2-distearoryl-sn-glycero-3-phosphatidyl choline (DSPC), sphingomyelin, egg phosphatidylcholines and monosialoganglioside. Since this formulation is made up of phospholipids only, liposomal formulations have encountered many challenges, one of the ones being the instability in plasma. Several attempts to overcome these challenges have been made, specifically in the manipulation of the lipid membrane. One of these attempts focused on the manipulation of cholesterol. Addition of cholesterol to conventional formulations reduces rapid release of the encapsulated bioactive compound into the plasma or 1,2-dioleoyl-sn-glycero-3-phosphoethanolamine (DOPE) increases the stability (see, e.g., Spuch and Navarro, Journal of Drug Delivery, vol. 2011, Article ID 469679, 12 pages, 2011. doi:10.1155/2011/469679 for review).

[00502] In a particularly advantageous embodiment, Trojan Horse liposomes (also known as Molecular Trojan Horses) are desirable and protocols may be found at http://cshprotocols.cshlp.org/content/2010/4/pdb.prot5407.long. These particles allow delivery of a transgene to the entire brain after an intravascular injection. Without being bound by limitation, it is believed that neutral lipid particles with specific antibodies conjugated to surface allow crossing of the blood brain barrier via endocytosis. Trojan Horse Liposomes may be used to deliver the CRISPR family of nucleases to the brain via an intravascular injection, which would allow whole brain transgenic animals without the need for embryonic manipulation. About 1-5 g of DNA or RNA may be contemplated for in vivo administration in liposomes.

[00503] In another embodiment, the AD-functionalized CRISPR Cas system or components thereof may be administered in liposomes, such as a stable nucleic-acid-lipid particle (SNALP) (see, e.g., Morrissey et al., Nature Biotechnology, Vol. 23, No. 8, August 2005). Daily intravenous injections of about 1, 3 or 5 mg/kg/day of a specific CRISPR Cas targeted in a SNALP are contemplated. The daily treatment may be over about three days

and then weekly for about five weeks. In another embodiment, a specific CRISPR Cas encapsulated SNALP) administered by intravenous injection to at doses of about 1 or 2.5 mg/kg are also contemplated (see, e.g., Zimmerman et al., Nature Letters, Vol. 441, 4 May 2006). The SNALP formulation may contain the lipids 3-N-[(wmethoxypoly(ethylene glycol) 2000) carbamoyl] -1,2-dimyristyloxy-propylamine (PEG-C-DMA), 1,2-dilinoleyloxy-N,N-dimethyl-3-aminopropane (DLinDMA), 1,2-distearoyl-sn-glycero-3-phosphocholine (DSPC) and cholesterol, in a 2:40:10:48 molar per cent ratio (see, e.g., Zimmerman et al., Nature Letters, Vol. 441, 4 May 2006).

[00504] In another embodiment, stable nucleic-acid-lipid particles (SNALPs) have proven to be effective delivery molecules to highly vascularized HepG2-derived liver tumors but not in poorly vascularized HCT-116 derived liver tumors (see, e.g., Li, Gene Therapy (2012) 19, 775–780). The SNALP liposomes may be prepared by formulating D-Lin-DMA and PEG-C-DMA with distearoylphosphatidylcholine (DSPC), Cholesterol and siRNA using a 25:1 lipid/siRNA ratio and a 48/40/10/2 molar ratio of Cholesterol/D-Lin-DMA/DSPC/PEG-C-DMA. The resulted SNALP liposomes are about 80–100 nm in size.

[00505] In yet another embodiment, a SNALP may comprise synthetic cholesterol (Sigma-Aldrich, St Louis, MO, USA), dipalmitoylphosphatidylcholine (Avanti Polar Lipids, Alabaster, AL, USA), 3-N-[(w-methoxy poly(ethylene glycol)2000)carbamoyl]-1,2-dimyrestyloxypropylamine, and cationic 1,2-dilinoleyloxy-3-N,Ndimethylaminopropane (see, e.g., Geisbert et al., Lancet 2010; 375: 1896-905). A dosage of about 2 mg/kg total CRISPR Cas per dose administered as, for example, a bolus intravenous infusion may be contemplated.

[00506] In yet another embodiment, a SNALP may comprise synthetic cholesterol (Sigma-Aldrich), 1,2-distearoyl-sn-glycero-3-phosphocholine (DSPC; Avanti Polar Lipids Inc.), PEG-cDMA, and 1,2-dilinoleyloxy-3-(N;N-dimethyl)aminopropane (DLinDMA) (see, e.g., Judge, J. Clin. Invest. 119:661-673 (2009)). Formulations used for in vivo studies may comprise a final lipid/RNA mass ratio of about 9:1.

[00507] The safety profile of RNAi nanomedicines has been reviewed by Barros and Gollob of Alnylam Pharmaceuticals (see, e.g., Advanced Drug Delivery Reviews 64 (2012) 1730–1737). The stable nucleic acid lipid particle (SNALP) is comprised of four different lipids — an ionizable lipid (DLinDMA) that is cationic at low pH, a neutral helper lipid, cholesterol, and a diffusible polyethylene glycol (PEG)-lipid. The particle is approximately 80 nm in diameter and is charge-neutral at physiologic pH. During formulation, the ionizable lipid serves to condense lipid with the anionic RNA during particle formation.

When positively charged under increasingly acidic endosomal conditions, the ionizable lipid also mediates the fusion of SNALP with the endosomal membrane enabling release of RNA into the cytoplasm. The PEG-lipid stabilizes the particle and reduces aggregation during formulation, and subsequently provides a neutral hydrophilic exterior that improves pharmacokinetic properties.

[00508] To date, two clinical programs have been initiated using SNALP formulations with RNA. Tekmira Pharmaceuticals recently completed a phase I single-dose study of SNALP-ApoB in adult volunteers with elevated LDL cholesterol. ApoB is predominantly expressed in the liver and jejunum and is essential for the assembly and secretion of VLDL and LDL. Seventeen subjects received a single dose of SNALP-ApoB (dose escalation across 7 dose levels). There was no evidence of liver toxicity (anticipated as the potential dose-limiting toxicity based on preclinical studies). One (of two) subjects at the highest dose experienced flu-like symptoms consistent with immune system stimulation, and the decision was made to conclude the trial.

[00509] Alnylam Pharmaceuticals has similarly advanced ALN-TTR01, which employs the SNALP technology described above and targets hepatocyte production of both mutant and wild-type TTR to treat TTR amyloidosis (ATTR). Three ATTR syndromes have been described: familial amyloidotic polyneuropathy (FAP) and familial amyloidotic cardiomyopathy (FAC) — both caused by autosomal dominant mutations in TTR; and senile systemic amyloidosis (SSA) cause by wildtype TTR. A placebo-controlled, single dose-escalation phase I trial of ALN-TTR01 was recently completed in patients with ATTR. ALN-TTR01 was administered as a 15-minute IV infusion to 31 patients (23 with study drug and 8 with placebo) within a dose range of 0.01 to 1.0 mg/kg (based on siRNA). Treatment was well tolerated with no significant increases in liver function tests. Infusion-related reactions were noted in 3 of 23 patients at≥0.4 mg/kg; all responded to slowing of the infusion rate and all continued on study. Minimal and transient elevations of serum cytokines IL-6, IP-10 and IL-1ra were noted in two patients at the highest dose of 1 mg/kg (as anticipated from preclinical and NHP studies). Lowering of serum TTR, the expected pharmacodynamics effect of ALN-TTR01, was observed at 1 mg/kg.

[00510] In yet another embodiment, a SNALP may be made by solubilizing a cationic lipid, DSPC, cholesterol and PEG-lipid e.g., in ethanol, e.g., at a molar ratio of 40:10:40:10, respectively (see, Semple et al., Nature Niotechnology, Volume 28 Number 2 February 2010, pp. 172-177). The lipid mixture was added to an aqueous buffer (50 mM citrate, pH 4) with mixing to a final ethanol and lipid concentration of 30% (vol/vol) and 6.1 mg/ml,

respectively, and allowed to equilibrate at 22 °C for 2 min before extrusion. The hydrated lipids were extruded through two stacked 80 nm pore-sized filters (Nuclepore) at 22 °C using a Lipex Extruder (Northern Lipids) until a vesicle diameter of 70–90 nm, as determined by dynamic light scattering analysis, was obtained. This generally required 1–3 passes. The siRNA (solubilized in a 50 mM citrate, pH 4 aqueous solution containing 30% ethanol) was added to the pre-equilibrated (35 °C) vesicles at a rate of ~5 ml/min with mixing. After a final target siRNA/lipid ratio of 0.06 (wt/wt) was reached, the mixture was incubated for a further 30 min at 35 °C to allow vesicle reorganization and encapsulation of the siRNA. The ethanol was then removed and the external buffer replaced with PBS (155 mM NaCl, 3 mM Na_2HPO_4, 1 mM KH_2PO_4, pH 7.5) by either dialysis or tangential flow diafiltration. siRNA were encapsulated in SNALP using a controlled step-wise dilution method process. The lipid constituents of KC2-SNALP were DLin-KC2-DMA (cationic lipid), dipalmitoylphosphatidylcholine (DPPC; Avanti Polar Lipids), synthetic cholesterol (Sigma) and PEG-C-DMA used at a molar ratio of 57.1:7.1:34.3:1.4. Upon formation of the loaded particles, SNALP were dialyzed against PBS and filter sterilized through a 0.2 μm filter before use. Mean particle sizes were 75–85 nm and 90–95% of the siRNA was encapsulated within the lipid particles. The final siRNA/lipid ratio in formulations used for in vivo testing was ~0.15 (wt/wt). LNP-siRNA systems containing Factor VII siRNA were diluted to the appropriate concentrations in sterile PBS immediately before use and the formulations were administered intravenously through the lateral tail vein in a total volume of 10 ml/kg. This method and these delivery systems may be extrapolated to the AD-functionalized CRISPR Cas system of the present invention.

[00511] Other Lipids

[00512] Other cationic lipids, such as amino lipid 2,2-dilinoleyl-4-dimethylaminoethyl-[1,3]-dioxolane (DLin-KC2-DMA) may be utilized to encapsulate CRISPR Cas or components thereof or nucleic acid molecule(s) coding therefor e.g., similar to SiRNA (see, e.g., Jayaraman, Angew. Chem. Int. Ed. 2012, 51, 8529 –8533), and hence may be employed in the practice of the invention. A preformed vesicle with the following lipid composition may be contemplated: amino lipid, distearoylphosphatidylcholine (DSPC), cholesterol and (R)-2,3-bis(octadecyloxy) propyl-1-(methoxy poly(ethylene glycol)2000)propylcarbamate (PEG-lipid) in the molar ratio 40/10/40/10, respectively, and a FVII siRNA/total lipid ratio of approximately 0.05 (w/w). To ensure a narrow particle size distribution in the range of 70–90 nm and a low polydispersity index of 0.11±0.04 (n=56), the particles may be extruded up to three times through 80 nm membranes prior to adding

the guide RNA. Particles containing the highly potent amino lipid 16 may be used, in which the molar ratio of the four lipid components 16, DSPC, cholesterol and PEG-lipid (50/10/38.5/1.5) which may be further optimized to enhance in vivo activity.

[00513] Michael S D Kormann et al. ("Expression of therapeutic proteins after delivery of chemically modified mRNA in mice: Nature Biotechnology, Volume:29, Pages: 154–157 (2011)) describes the use of lipid envelopes to deliver RNA. Use of lipid envelopes is also preferred in the present invention.

[00514] In another embodiment, lipids may be formulated with the AD-functionalized CRISPR Cas system of the present invention or component(s) thereof or nucleic acid molecule(s) coding therefor to form lipid nanoparticles (LNPs). Lipids include, but are not limited to, DLin-KC2-DMA4, C12-200 and colipids disteroylphosphatidyl choline, cholesterol, and PEG-DMG may be formulated with CRISPR Cas instead of siRNA (see, e.g., Novobrantseva, Molecular Therapy–Nucleic Acids (2012) 1, e4; doi:10.1038/mtna.2011.3) using a spontaneous vesicle formation procedure. The component molar ratio may be about 50/10/38.5/1.5 (DLin-KC2-DMA or C12-200/disteroylphosphatidyl choline/cholesterol/PEG-DMG). The final lipid:siRNA weight ratio may be ~12:1 and 9:1 in the case of DLin-KC2-DMA and C12-200 lipid nanoparticles (LNPs), respectively. The formulations may have mean particle diameters of ~80 nm with >90% entrapment efficiency. A 3 mg/kg dose may be contemplated.

[00515] Tekmira has a portfolio of approximately 95 patent families, in the U.S. and abroad, that are directed to various aspects of LNPs and LNP formulations (see, e.g., U.S. Pat. Nos. 7,982,027; 7,799,565; 8,058,069; 8,283,333; 7,901,708; 7,745,651; 7,803,397; 8,101,741; 8,188,263; 7,915,399; 8,236,943 and 7,838,658 and European Pat. Nos 1766035; 1519714; 1781593 and 1664316), all of which may be used and/or adapted to the present invention.

[00516] The AD-functionalized CRISPR Cas system or components thereof or nucleic acid molecule(s) coding therefor may be delivered encapsulated in PLGA Microspheres such as that further described in US published applications 20130252281 and 20130245107 and 20130244279 (assigned to Moderna Therapeutics) which relate to aspects of formulation of compositions comprising modified nucleic acid molecules which may encode a protein, a protein precursor, or a partially or fully processed form of the protein or a protein precursor. The formulation may have a molar ratio 50:10:38.5:1.5-3.0 (cationic lipid:fusogenic lipid:cholesterol:PEG lipid). The PEG lipid may be selected from, but is not limited to PEG-c-DOMG, PEG-DMG. The fusogenic lipid may be DSPC. See also,

Schrum et al., Delivery and Formulation of Engineered Nucleic Acids, US published application 20120251618.

[00517] Nanomerics' technology addresses bioavailability challenges for a broad range of therapeutics, including low molecular weight hydrophobic drugs, peptides, and nucleic acid based therapeutics (plasmid, siRNA, miRNA). Specific administration routes for which the technology has demonstrated clear advantages include the oral route, transport across the blood-brain-barrier, delivery to solid tumours, as well as to the eye. See, e.g., Mazza et al., 2013, ACS Nano. 2013 Feb 26;7(2):1016-26; Uchegbu and Siew, 2013, J Pharm Sci. 102(2):305-10 and Lalatsa et al., 2012, J Control Release. 2012 Jul 20; 161(2):523-36.

[00518] US Patent Publication No. 20050019923 describes cationic dendrimers for delivering bioactive molecules, such as polynucleotide molecules, peptides and polypeptides and/or pharmaceutical agents, to a mammalian body. The dendrimers are suitable for targeting the delivery of the bioactive molecules to, for example, the liver, spleen, lung, kidney or heart (or even the brain). Dendrimers are synthetic 3-dimensional macromolecules that are prepared in a step-wise fashion from simple branched monomer units, the nature and functionality of which can be easily controlled and varied. Dendrimers are synthesised from the repeated addition of building blocks to a multifunctional core (divergent approach to synthesis), or towards a multifunctional core (convergent approach to synthesis) and each addition of a 3-dimensional shell of building blocks leads to the formation of a higher generation of the dendrimers. Polypropylenimine dendrimers start from a diaminobutane core to which is added twice the number of amino groups by a double Michael addition of acrylonitrile to the primary amines followed by the hydrogenation of the nitriles. This results in a doubling of the amino groups. Polypropylenimine dendrimers contain 100% protonable nitrogens and up to 64 terminal amino groups (generation 5, DAB 64). Protonable groups are usually amine groups which are able to accept protons at neutral pH. The use of dendrimers as gene delivery agents has largely focused on the use of the polyamidoamine. and phosphorous containing compounds with a mixture of amine/amide or N--P(O_2)S as the conjugating units respectively with no work being reported on the use of the lower generation polypropylenimine dendrimers for gene delivery. Polypropylenimine dendrimers have also been studied as pH sensitive controlled release systems for drug delivery and for their encapsulation of guest molecules when chemically modified by peripheral amino acid groups. The cytotoxicity and interaction of polypropylenimine dendrimers with DNA as well as the transfection efficacy of DAB 64 has also been studied.

[00519] US Patent Publication No. 20050019923 is based upon the observation that, contrary to earlier reports, cationic dendrimers, such as polypropylenimine dendrimers, display suitable properties, such as specific targeting and low toxicity, for use in the targeted delivery of bioactive molecules, such as genetic material. In addition, derivatives of the cationic dendrimer also display suitable properties for the targeted delivery of bioactive molecules. See also, Bioactive Polymers, US published application 20080267903, which discloses "Various polymers, including cationic polyamine polymers and dendrimeric polymers, are shown to possess anti-proliferative activity, and may therefore be useful for treatment of disorders characterised by undesirable cellular proliferation such as neoplasms and tumours, inflammatory disorders (including autoimmune disorders), psoriasis and atherosclerosis. The polymers may be used alone as active agents, or as delivery vehicles for other therapeutic agents, such as drug molecules or nucleic acids for gene therapy. In such cases, the polymers' own intrinsic anti-tumour activity may complement the activity of the agent to be delivered." The disclosures of these patent publications may be employed in conjunction with herein teachings for delivery of AD-functionalized CRISPR Cas system(s) or component(s) thereof or nucleic acid molecule(s) coding therefor.

[00520] Supercharged proteins

[00521] Supercharged proteins are a class of engineered or naturally occurring proteins with unusually high positive or negative net theoretical charge and may be employed in delivery of AD-functionalized CRISPR Cas system(s) or component(s) thereof or nucleic acid molecule(s) coding therefor. Both supernegatively and superpositively charged proteins exhibit a remarkable ability to withstand thermally or chemically induced aggregation. Superpositively charged proteins are also able to penetrate mammalian cells. Associating cargo with these proteins, such as plasmid DNA, RNA, or other proteins, can enable the functional delivery of these macromolecules into mammalian cells both in vitro and in vivo. The creation and characterization of supercharged proteins has been reported in 2007 (Lawrence et al., 2007, Journal of the American Chemical Society 129, 10110–10112).

[00522] The nonviral delivery of RNA and plasmid DNA into mammalian cells are valuable both for research and therapeutic applications (Akinc et al., 2010, Nat. Biotech. 26, 561–569). Purified +36 GFP protein (or other superpositively charged protein) is mixed with RNAs in the appropriate serum-free media and allowed to complex prior addition to cells. Inclusion of serum at this stage inhibits formation of the supercharged protein-RNA complexes and reduces the effectiveness of the treatment. The following protocol has been found to be effective for a variety of cell lines (McNaughton et al., 2009, Proc. Natl. Acad.

Sci. USA 106, 6111–6116) (However, pilot experiments varying the dose of protein and RNA should be performed to optimize the procedure for specific cell lines): (1) One day before treatment, plate 1 x 10^5 cells per well in a 48-well plate. (2) On the day of treatment, dilute purified +36 GFP protein in serumfree media to a final concentration 200nM. Add RNA to a final concentration of 50nM. Vortex to mix and incubate at room temperature for 10min. (3) During incubation, aspirate media from cells and wash once with PBS. (4) Following incubation of +36 GFP and RNA, add the protein-RNA complexes to cells. (5) Incubate cells with complexes at 37 °C for 4h. (6) Following incubation, aspirate the media and wash three times with 20 U/mL heparin PBS. Incubate cells with serum-containing media for a further 48h or longer depending upon the assay for activity. (7) Analyze cells by immunoblot, qPCR, phenotypic assay, or other appropriate method.

[00523] It has been further found +36 GFP to be an effective plasmid delivery reagent in a range of cells. As plasmid DNA is a larger cargo than siRNA, proportionately more +36 GFP protein is required to effectively complex plasmids. For effective plasmid delivery Applicants have developed a variant of +36 GFP bearing a C-terminal HA2 peptide tag, a known endosome-disrupting peptide derived from the influenza virus hemagglutinin protein. The following protocol has been effective in a variety of cells, but as above it is advised that plasmid DNA and supercharged protein doses be optimized for specific cell lines and delivery applications: (1) One day before treatment, plate 1 x 10^5 per well in a 48-well plate. (2) On the day of treatment, dilute purified þ36 GFP protein in serumfree media to a final concentration 2 mM. Add 1mg of plasmid DNA. Vortex to mix and incubate at room temperature for 10min. (3) During incubation, aspirate media from cells and wash once with PBS. (4) Following incubation of þ36 GFP and plasmid DNA, gently add the protein-DNA complexes to cells. (5) Incubate cells with complexes at 37 C for 4h. (6) Following incubation, aspirate the media and wash with PBS. Incubate cells in serum-containing media and incubate for a further 24–48h. (7) Analyze plasmid delivery (e.g., by plasmid-driven gene expression) as appropriate.

[00524] See also, e.g., McNaughton et al., Proc. Natl. Acad. Sci. USA 106, 6111-6116 (2009); Cronican et al., ACS Chemical Biology 5, 747-752 (2010); Cronican et al., Chemistry & Biology 18, 833-838 (2011); Thompson et al., Methods in Enzymology 503, 293-319 (2012); Thompson, D.B., et al., Chemistry & Biology 19 (7), 831-843 (2012). The methods of the super charged proteins may be used and/or adapted for delivery of the AD-functionalized CRISPR Cas system of the present invention. These systems in conjunction

with herein teaching can be employed in the delivery of AD-functionalized CRISPR Cas system(s) or component(s) thereof or nucleic acid molecule(s) coding therefor

[00525] Cell Penetrating Peptides (CPPs)

[00526] In yet another embodiment, cell penetrating peptides (CPPs) are contemplated for the delivery of the AD-functionalized CRISPR Cas system. CPPs are short peptides that facilitate cellular uptake of various molecular cargo (from nanosize particles to small chemical molecules and large fragments of DNA). The term "cargo" as used herein includes but is not limited to the group consisting of therapeutic agents, diagnostic probes, peptides, nucleic acids, antisense oligonucleotides, plasmids, proteins, particles, including nanoparticles, liposomes, chromophores, small molecules and radioactive materials. In aspects of the invention, the cargo may also comprise any component of the AD-functionalized CRISPR Cas system or the entire AD-functionalized functional CRISPR Cas system. Aspects of the present invention further provide methods for delivering a desired cargo into a subject comprising: (a) preparing a complex comprising the cell penetrating peptide of the present invention and a desired cargo, and (b) orally, intraarticularly, intraperitoneally, intrathecally, intrarterially, intranasally, intraparenchymally, subcutaneously, intramuscularly, intravenously, dermally, intrarectally, or topically administering the complex to a subject. The cargo is associated with the peptides either through chemical linkage via covalent bonds or through non-covalent interactions.

[00527] The function of the CPPs are to deliver the cargo into cells, a process that commonly occurs through endocytosis with the cargo delivered to the endosomes of living mammalian cells. Cell-penetrating peptides are of different sizes, amino acid sequences, and charges but all CPPs have one distinct characteristic, which is the ability to translocate the plasma membrane and facilitate the delivery of various molecular cargoes to the cytoplasm or an organelle. CPP translocation may be classified into three main entry mechanisms: direct penetration in the membrane, endocytosis-mediated entry, and translocation through the formation of a transitory structure. CPPs have found numerous applications in medicine as drug delivery agents in the treatment of different diseases including cancer and virus inhibitors, as well as contrast agents for cell labeling. Examples of the latter include acting as a carrier for GFP, MRI contrast agents, or quantum dots. CPPs hold great potential as in vitro and in vivo delivery vectors for use in research and medicine. CPPs typically have an amino acid composition that either contains a high relative abundance of positively charged amino acids such as lysine or arginine or has sequences that contain an alternating pattern of polar/charged amino acids and non-polar, hydrophobic amino acids. These two types of

structures are referred to as polycationic or amphipathic, respectively. A third class of CPPs are the hydrophobic peptides, containing only apolar residues, with low net charge or have hydrophobic amino acid groups that are crucial for cellular uptake. One of the initial CPPs discovered was the trans-activating transcriptional activator (Tat) from Human Immunodeficiency Virus 1 (HIV-1) which was found to be efficiently taken up from the surrounding media by numerous cell types in culture. Since then, the number of known CPPs has expanded considerably and small molecule synthetic analogues with more effective protein transduction properties have been generated. CPPs include but are not limited to Penetratin, Tat (48-60), Transportan, and (R-AhX-R4) (Ahx=aminohexanoyl).

[00528] US Patent 8,372,951, provides a CPP derived from eosinophil cationic protein (ECP) which exhibits highly cell-penetrating efficiency and low toxicity. Aspects of delivering the CPP with its cargo into a vertebrate subject are also provided. Further aspects of CPPs and their delivery are described in U. S. patents 8,575,305; 8;614,194 and 8,044,019. CPPs can be used to deliver the AD-functionalized CRISPR-Cas system or components thereof. That CPPs can be employed to deliver the AD-functionalized CRISPR-Cas system or components thereof is also provided in the manuscript "Gene disruption by cell-penetrating peptide-mediated delivery of Cas9 protein and guide RNA", by Suresh Ramakrishna, Abu-Bonsrah Kwaku Dad, Jagadish Beloor, et al. Genome Res. 2014 Apr 2, incorporated by reference in its entirety, wherein it is demonstrated that treatment with CPP-conjugated recombinant Cas9 protein and CPP-complexed guide RNAs lead to endogenous gene disruptions in human cell lines. In the paper the Cas9 protein was conjugated to CPP via a thioether bond, whereas the guide RNA was complexed with CPP, forming condensed, positively charged particles. It was shown that simultaneous and sequential treatment of human cells, including embryonic stem cells, dermal fibroblasts, HEK293T cells, HeLa cells, and embryonic carcinoma cells, with the modified Cas9 and guide RNA led to efficient gene disruptions with reduced off-target mutations relative to plasmid transfections.

[00529] **Aerosol delivery**

[00530] Subjects treated for a lung disease may for example receive pharmaceutically effective amount of aerosolized AAV vector system per lung endobronchially delivered while spontaneously breathing. As such, aerosolized delivery is preferred for AAV delivery in general. An adenovirus or an AAV particle may be used for delivery. Suitable gene constructs, each operably linked to one or more regulatory sequences, may be cloned into the delivery vector.

[00531] Packaging and Promoters

[00532] The promoter used to drive CRISPR-Cas protein and adenosine deaminase coding nucleic acid molecule expression can include AAV ITR, which can serve as a promoter. This is advantageous for eliminating the need for an additional promoter element (which can take up space in the vector). The additional space freed up can be used to drive the expression of additional elements (gRNA, etc.). Also, ITR activity is relatively weaker, so can be used to reduce potential toxicity due to over expression of Cpf1.

[00533] For ubiquitous expression, promoters that can be used include: CMV, CAG, CBh, PGK, SV40, Ferritin heavy or light chains, etc. For brain or other CNS expression, SynapsinI can be used for all neurons, CaMKIIalpha can be used for excitatory neurons, GAD67 or GAD65 or VGAT can be used for GABAergic neurons. For liver expression, Albumin promoter can be used. For lung expression, SP-B can be used. For endothelial cells, ICAM can be used. For hematopoietic cells, IFNbeta or CD45 can be used. For Osteoblasts, the OG-2 can be used.

[00534] The promoter used to drive guide RNA can include Pol III promoters such as U6 or H1, as well as use of Pol II promoter and intronic cassettes to express guide RNA.

[00535] Adeno associated virus (AAV)

[00536] The CRISPR-Cas protein, adenosine deaminase, and one or more guide RNA can be delivered using adeno associated virus (AAV), lentivirus, adenovirus or other plasmid or viral vector types, in particular, using formulations and doses from, for example, US Patents Nos. 8,454,972 (formulations, doses for adenovirus), 8,404,658 (formulations, doses for AAV) and 5,846,946 (formulations, doses for DNA plasmids) and from clinical trials and publications regarding the clinical trials involving lentivirus, AAV and adenovirus. For examples, for AAV, the route of administration, formulation and dose can be as in US Patent No. 8,454,972 and as in clinical trials involving AAV. For Adenovirus, the route of administration, formulation and dose can be as in US Patent No. 8,404,658 and as in clinical trials involving adenovirus. For plasmid delivery, the route of administration, formulation and dose can be as in US Patent No 5,846,946 and as in clinical studies involving plasmids. Doses may be based on or extrapolated to an average 70 kg individual (e.g. a male adult human), and can be adjusted for patients, subjects, mammals of different weight and species. Frequency of administration is within the ambit of the medical or veterinary practitioner (e.g., physician, veterinarian), depending on usual factors including the age, sex, general health, other conditions of the patient or subject and the particular condition or symptoms being addressed. The viral vectors can be injected into the tissue of

interest. For cell-type specific genome modification, the expression of Cpf1 and adenosine deaminase can be driven by a cell-type specific promoter. For example, liver-specific expression might use the Albumin promoter and neuron-specific expression (e.g. for targeting CNS disorders) might use the Synapsin I promoter.

[00537] In terms of in vivo delivery, AAV is advantageous over other viral vectors for a couple of reasons: low toxicity (this may be due to the purification method not requiring ultra centrifugation of cell particles that can activate the immune response); and low probability of causing insertional mutagenesis because it doesn't integrate into the host genome.

[00538] AAV has a packaging limit of 4.5 or 4.75 Kb. This means that Cpf1 as well as a promoter and transcription terminator have to be all fit into the same viral vector. Constructs larger than 4.5 or 4.75 Kb will lead to significantly reduced virus production. SpCas9 is quite large, the gene itself is over 4.1 Kb, which makes it difficult for packing into AAV. Therefore embodiments of the invention include utilizing homologs of Cpf1 that are shorter.

[00539] As to AAV, the AAV can be AAV1, AAV2, AAV5 or any combination thereof. One can select the AAV of the AAV with regard to the cells to be targeted; e.g., one can select AAV serotypes 1, 2, 5 or a hybrid capsid AAV1, AAV2, AAV5 or any combination thereof for targeting brain or neuronal cells; and one can select AAV4 for targeting cardiac tissue. AAV8 is useful for delivery to the liver. The herein promoters and vectors are preferred individually. A tabulation of certain AAV serotypes as to these cells (see Grimm, D. et al, J. Virol. 82: 5887-5911 (2008)) is as follows:

Cell Line	AAV-1	AAV-2	AAV-3	AAV-4	AAV-5	AAV-6	AAV-8	AAV-9
Huh-7	13	100	2.5	0.0	0.1	10	0.7	0.0
HEK293	25	100	2.5	0.1	0.1	5	0.7	0.1
HeLa	3	100	2.0	0.1	6.7	1	0.2	0.1
HepG2	3	100	16.7	0.3	1.7	5	0.3	ND
Hep1A	20	100	0.2	1.0	0.1	1	0.2	0.0
911	17	100	11	0.2	0.1	17	0.1	ND
CHO	100	100	14	1.4	333	50	10	1.0
COS	33	100	33	3.3	5.0	14	2.0	0.5
MeWo	10	100	20	0.3	6.7	10	1.0	0.2
NIH3T3	10	100	2.9	2.9	0.3	10	0.3	ND
A549	14	100	20	ND	0.5	10	0.5	0.1
HT1180	20	100	10	0.1	0.3	33	0.5	0.1
Monocytes	1111	100	ND	ND	125	1429	ND	ND
Immature	2500	100	ND	ND	222	2857	ND	ND

DC								
Mature DC	2222	100	ND	ND	333	3333	ND	ND

[00540] Lentiviruses

[00541] Lentiviruses are complex retroviruses that have the ability to infect and express their genes in both mitotic and post-mitotic cells. The most commonly known lentivirus is the human immunodeficiency virus (HIV), which uses the envelope glycoproteins of other viruses to target a broad range of cell types.

[00542] Lentiviruses may be prepared as follows. After cloning pCasES10 (which contains a lentiviral transfer plasmid backbone), HEK293FT at low passage (p=5) were seeded in a T-75 flask to 50% confluence the day before transfection in DMEM with 10% fetal bovine serum and without antibiotics. After 20 hours, media was changed to OptiMEM (serum-free) media and transfection was done 4 hours later. Cells were transfected with 10 μg of lentiviral transfer plasmid (pCasES10) and the following packaging plasmids: 5 μg of pMD2.G (VSV-g pseudotype), and 7.5ug of psPAX2 (gag/pol/rev/tat). Transfection was done in 4mL OptiMEM with a cationic lipid delivery agent (50uL Lipofectamine 2000 and 100ul Plus reagent). After 6 hours, the media was changed to antibiotic-free DMEM with 10% fetal bovine serum. These methods use serum during cell culture, but serum-free methods are preferred.

[00543] Lentivirus may be purified as follows. Viral supernatants were harvested after 48 hours. Supernatants were first cleared of debris and filtered through a 0.45um low protein binding (PVDF) filter. They were then spun in a ultracentrifuge for 2 hours at 24,000 rpm. Viral pellets were resuspended in 50ul of DMEM overnight at 4C. They were then aliquotted and immediately frozen at -80°C.

[00544] In another embodiment, minimal non-primate lentiviral vectors based on the equine infectious anemia virus (EIAV) are also contemplated, especially for ocular gene therapy (see, e.g., Balagaan, J Gene Med 2006; 8: 275 – 285). In another embodiment, RetinoStat®, an equine infectious anemia virus-based lentiviral gene therapy vector that expresses angiostatic proteins endostatin and angiostatin that is delivered via a subretinal injection for the treatment of the web form of age-related macular degeneration is also contemplated (see, e.g., Binley et al., HUMAN GENE THERAPY 23:980–991 (September 2012)) and this vector may be modified for the AD-functionalized CRISPR-Cas system of the present invention.

[00545] In another embodiment, self-inactivating lentiviral vectors with an siRNA targeting a common exon shared by HIV tat/rev, a nucleolar-localizing TAR decoy, and an anti–CCR5-specific hammerhead ribozyme (see, e.g., DiGiusto et al. (2010) Sci Transl Med 2:36ra43) may be used/and or adapted to the AD-functionalized CRISPR-Cas system of the present invention. A minimum of 2.5 × 106 CD34+ cells per kilogram patient weight may be collected and prestimulated for 16 to 20 hours in X-VIVO 15 medium (Lonza) containing 2 μmol/L-glutamine, stem cell factor (100 ng/ml), Flt-3 ligand (Flt-3L) (100 ng/ml), and thrombopoietin (10 ng/ml) (CellGenix) at a density of 2 × 106 cells/ml. Prestimulated cells may be transduced with lentiviral at a multiplicity of infection of 5 for 16 to 24 hours in 75-cm2 tissue culture flasks coated with fibronectin (25 mg/cm2) (RetroNectin,Takara Bio Inc.).

[00546] Lentiviral vectors have been disclosed as in the treatment for Parkinson's Disease, see, e.g., US Patent Publication No. 20120295960 and US Patent Nos. 7303910 and 7351585. Lentiviral vectors have also been disclosed for the treatment of ocular diseases, see e.g., US Patent Publication Nos. 20060281180, 20090007284, US20110117189; US20090017543; US20070054961, US20100317109. Lentiviral vectors have also been disclosed for delivery to the brain, see, e.g., US Patent Publication Nos. US20110293571; US20110293571, US20040013648, US20070025970, US20090111106 and US Patent No. US7259015.

[00547] Application in Non-Animal Organisms

[00548] The AD-functionalized CRISPR system(s) (e.g., single or multiplexed) can be used in conjunction with recent advances in crop genomics. The systems described herein can be used to perform efficient and cost effective plant gene or genome interrogation or editing or manipulation—for instance, for rapid investigation and/or selection and/or interrogations and/or comparison and/or manipulations and/or transformation of plant genes or genomes; e.g., to create, identify, develop, optimize, or confer trait(s) or characteristic(s) to plant(s) or to transform a plant genome. There can accordingly be improved production of plants, new plants with new combinations of traits or characteristics or new plants with enhanced traits. The AD-functionalized CRISPR system can be used with regard to plants in Site-Directed Integration (SDI) or Gene Editing (GE) or any Near Reverse Breeding (NRB) or Reverse Breeding (RB) techniques. Aspects of utilizing the herein described Cpf1 effector protein systems may be analogous to the use of the CRISPR-Cas (e.g. CRISPR-Cas9) system in plants, and mention is made of the University of Arizona website "CRISPR-PLANT" (http://www.genome.arizona.edu/crispr/) (supported by Penn State and

AGI). Emodiments of the invention can be used in genome editing in plants or where RNAi or similar genome editing techniques have been used previously; see, e.g., Nekrasov, "Plant genome editing made easy: targeted mutagenesis in model and crop plants using the CRISPR-Cas system," Plant Methods 2013, 9:39 (doi:10.1186/1746-4811-9-39); Brooks, "Efficient gene editing in tomato in the first generation using the CRISPR-Cas9 system," Plant Physiology September 2014 pp 114.247577; Shan, "Targeted genome modification of crop plants using a CRISPR-Cas system," Nature Biotechnology 31, 686-688 (2013); Feng, "Efficient genome editing in plants using a CRISPR-Cas system," Cell Research (2013) 23:1229–1232. doi:10.1038/cr.2013.114; published online 20 August 2013; Xie, "RNA-guided genome editing in plants using a CRISPR-Cas system," Mol Plant. 2013 Nov;6(6):1975-83. doi: 10.1093/mp/sst119. Epub 2013 Aug 17; Xu, "Gene targeting using the Agrobacterium tumefaciens-mediated CRISPR-Cas system in rice," Rice 2014, 7:5 (2014), Zhou et al., "Exploiting SNPs for biallelic CRISPR mutations in the outcrossing woody perennial Populus reveals 4-coumarate: CoA ligase specificity and Redundancy," New Phytologist (2015) (Forum) 1-4 (available online only at www.newphytologist.com); Caliando et al, "Targeted DNA degradation using a CRISPR device stably carried in the host genome, NATURE COMMUNICATIONS 6:6989, DOI: 10.1038/ncomms7989, www.nature.com/naturecommunications DOI: 10.1038/ncomms7989; US Patent No. 6,603,061 - Agrobacterium-Mediated Plant Transformation Method; US Patent No. 7,868,149 - Plant Genome Sequences and Uses Thereof and US 2009/0100536 - Transgenic Plants with Enhanced Agronomic Traits, all the contents and disclosure of each of which are herein incorporated by reference in their entirety. In the practice of the invention, the contents and disclosure of Morrell et al "Crop genomics: advances and applications," Nat Rev Genet. 2011 Dec 29;13(2):85-96; each of which is incorporated by reference herein including as to how herein embodiments may be used as to plants. Accordingly, reference herein to animal cells may also apply, mutatis mutandis, to plant cells unless otherwise apparent; and, the enzymes herein having reduced off-target effects and systems employing such enzymes can be used in plant applciations, including those mentioned herein.

[00549] Application of AD-functionalized CRISPR system to plants and yeast

[00550] In general, the term "plant" relates to any various photosynthetic, eukaryotic, unicellular or multicellular organism of the kingdom Plantae characteristically growing by cell division, containing chloroplasts, and having cell walls comprised of cellulose. The term plant encompasses monocotyledonous and dicotyledonous plants. Specifically, the plants are intended to comprise without limitation angiosperm and gymnosperm plants such

as acacia, alfalfa, amaranth, apple, apricot, artichoke, ash tree, asparagus, avocado, banana, barley, beans, beet, birch, beech, blackberry, blueberry, broccoli, Brussel's sprouts, cabbage, canola, cantaloupe, carrot, cassava, cauliflower, cedar, a cereal, celery, chestnut, cherry, Chinese cabbage, citrus, clementine, clover, coffee, corn, cotton, cowpea, cucumber, cypress, eggplant, elm, endive, eucalyptus, fennel, figs, fir, geranium, grape, grapefruit, groundnuts, ground cherry, gum hemlock, hickory, kale, kiwifruit, kohlrabi, larch, lettuce, leek, lemon, lime, locust, pine, maidenhair, maize, mango, maple, melon, millet, mushroom, mustard, nuts, oak, oats, oil palm, okra, onion, orange, an ornamental plant or flower or tree, papaya, palm, parsley, parsnip, pea, peach, peanut, pear, peat, pepper, persimmon, pigeon pea, pine, pineapple, plantain, plum, pomegranate, potato, pumpkin, radicchio, radish, rapeseed, raspberry, rice, rye, sorghum, safflower, sallow, soybean, spinach, spruce, squash, strawberry, sugar beet, sugarcane, sunflower, sweet potato, sweet corn, tangerine, tea, tobacco, tomato, trees, triticale, turf grasses, turnips, vine, walnut, watercress, watermelon, wheat, yams, yew, and zucchini. The term plant also encompasses Algae, which are mainly photoautotrophs unified primarily by their lack of roots, leaves and other organs that characterize higher plants.

[00551] The methods for genome editing using the AD-functionalized CRISPR system as described herein can be used to confer desired traits on essentially any plant. A wide variety of plants and plant cell systems may be engineered for the desired physiological and agronomic characteristics described herein using the nucleic acid constructs of the present disclosure and the various transformation methods mentioned above. In preferred embodiments, target plants and plant cells for engineering include, but are not limited to, those monocotyledonous and dicotyledonous plants, such as crops including grain crops (e.g., wheat, maize, rice, millet, barley), fruit crops (e.g., tomato, apple, pear, strawberry, orange), forage crops (e.g., alfalfa), root vegetable crops (e.g., carrot, potato, sugar beets, yam), leafy vegetable crops (e.g., lettuce, spinach); flowering plants (e.g., petunia, rose, chrysanthemum), conifers and pine trees (e.g., pine fir, spruce); plants used in phytoremediation (e.g., heavy metal accumulating plants); oil crops (e.g., sunflower, rape seed) and plants used for experimental purposes (e.g., Arabidopsis). Thus, the methods and systems can be used over a broad range of plants, such as for example with dicotyledonous plants belonging to the orders Magniolales, Illiciales, Laurales, Piperales, Aristochiales, Nymphaeales, Ranunculales, Papeverales, Sarraceniaceae, Trochodendrales, Hamamelidales, Eucomiales, Leitneriales, Myricales, Fagales, Casuarinales, Caryophyllales, Batales, Polygonales, Plumbaginales, Dilleniales, Theales, Malvales,

Urticales, Lecythidales, Violales, Salicales, Capparales, Ericales, Diapensales, Ebenales, Primulales, Rosales, Fabales, Podostemales, Haloragales, Myrtales, Cornales, Proteales, San tales, Rafflesiales, Celastrales, Euphorbiales, Rhamnales, Sapindales, Juglandales, Geraniales, Polygalales, Umbellales, Gentianales, Polemoniales, Lamiales, Plantaginales, Scrophulariales, Campanulales, Rubiales, Dipsacales, and Asterales; the methods and CRISPR-Cas systems can be used with monocotyledonous plants such as those belonging to the orders Alismatales, Hydrocharitales, Najadales, Triuridales, Commelinales, Eriocaulales, Restionales, Poales, Juncales, Cyperales, Typhales, Bromeliales, Zingiberales, Arecales, Cyclanthales, Pandanales, Arales, Lilliales, and Orchid ales, or with plants belonging to Gymnospermae, e.g those belonging to the orders Pinales, Ginkgoales, Cycadales, Araucariales, Cupressales and Gnetales.

[00552] The AD-functionalized CRISPR systems and methods of use described herein can be used over a broad range of plant species, included in the non-limitative list of dicot, monocot or gymnosperm genera hereunder: *Atropa, Alseodaphne, Anacardium, Arachis, Beilschmiedia, Brassica, Carthamus, Cocculus, Croton, Cucumis, Citrus, Citrullus, Capsicum, Catharanthus, Cocos, Coffea, Cucurbita, Daucus, Duguetia, Eschscholzia, Ficus, Fragaria, Glaucium, Glycine, Gossypium, Helianthus, Hevea, Hyoscyamus, Lactuca, Landolphia, Linum, Litsea, Lycopersicon, Lupinus, Manihot, Majorana, Malus, Medicago, Nicotiana, Olea, Parthenium, Papaver, Persea, Phaseolus, Pistacia, Pisum, Pyrus, Prunus, Raphanus, Ricinus, Senecio, Sinomenium, Stephania, Sinapis, Solanum, Theobroma, Trifolium, Trigonella, Vicia, Vinca, Vilis,* and *Vigna; and* the genera *Allium, Andropogon, Aragrostis, Asparagus, Avena, Cynodon, Elaeis, Festuca, Festulolium, Heterocallis, Hordeum, Lemna, Lolium, Musa, Oryza, Panicum, Pannesetum, Phleum, Poa, Secale, Sorghum, Triticum, Zea, Abies, Cunninghamia, Ephedra, Picea, Pinus,* and *Pseudotsuga.*

[00553] The AD-functionalized CRISPR systems and methods of use can also be used over a broad range of "algae" or "algae cells"; including for example algea selected from several eukaryotic phyla, including the Rhodophyta (red algae), Chlorophyta (green algae), Phaeophyta (brown algae), Bacillariophyta (diatoms), Eustigmatophyta and dinoflagellates as well as the prokaryotic phylum Cyanobacteria (blue-green algae). The term "algae" includes for example algae selected from : Amphora, Anabaena, Anikstrodesmis, Botryococcus, Chaetoceros, Chlamydomonas, Chlorella, Chlorococcum, Cyclotella, Cylindrotheca, Dunaliella, Emiliana, Euglena, Hematococcus, Isochrysis, Monochrysis, Monoraphidium, Nannochloris, Nannnochloropsis, Navicula, Nephrochloris, Nephroselmis, Nitzschia, Nodularia, Nostoc, Oochromonas, Oocystis, Oscillartoria, Pavlova,

Phaeodactylum, Playtmonas, Pleurochrysis, Porhyra, Pseudoanabaena, Pyramimonas, Stichococcus, Synechococcus, Synechocystis, Tetraselmis, Thalassiosira, and Trichodesmium.

[00554] A part of a plant, i.e., a "plant tissue" may be treated according to the methods of the present invention to produce an improved plant. Plant tissue also encompasses plant cells.The term “plant cell” as used herein refers to individual units of a living plant, either in an intact whole plant or in an isolated form grown in in vitro tissue cultures, on media or agar, in suspension in a growth media or buffer or as a part of higher organized unites, such as, for example, plant tissue, a plant organ, or a whole plant.

[00555] A “protoplast” refers to a plant cell that has had its protective cell wall completely or partially removed using, for example, mechanical or enzymatic means resulting in an intact biochemical competent unit of living plant that can reform their cell wall, proliferate and regenerate grow into a whole plant under proper growing conditions.

[00556] The term "transformation" broadly refers to the process by which a plant host is genetically modified by the introduction of DNA by means of *Agrobacteria* or one of a variety of chemical or physical methods. As used herein, the term "plant host" refers to plants, including any cells, tissues, organs, or progeny of the plants. Many suitable plant tissues or plant cells can be transformed and include, but are not limited to, protoplasts, somatic embryos, pollen, leaves, seedlings, stems, calli, stolons, microtubers, and shoots. A plant tissue also refers to any clone of such a plant, seed, progeny, propagule whether generated sexually or asexually, and descendents of any of these, such as cuttings or seed.

[00557] The term "transformed" as used herein, refers to a cell, tissue, organ, or organism into which a foreign DNA molecule, such as a construct, has been introduced. The introduced DNA molecule may be integrated into the genomic DNA of the recipient cell, tissue, organ, or organism such that the introduced DNA molecule is transmitted to the subsequent progeny. In these embodiments, the "transformed" or “transgenic” cell or plant may also include progeny of the cell or plant and progeny produced from a breeding program employing such a transformed plant as a parent in a cross and exhibiting an altered phenotype resulting from the presence of the introduced DNA molecule. Preferably, the transgenic plant is fertile and capable of transmitting the introduced DNA to progeny through sexual reproduction.

[00558] The term “progeny”, such as the progeny of a transgenic plant, is one that is born of, begotten by, or derived from a plant or the transgenic plant. The introduced DNA molecule may also be transiently introduced into the recipient cell such that the introduced

DNA molecule is not inherited by subsequent progeny and thus not considered "transgenic". Accordingly, as used herein, a "non-transgenic" plant or plant cell is a plant which does not contain a foreign DNA stably integrated into its genome.

[00559] The term "plant promoter" as used herein is a promoter capable of initiating transcription in plant cells, whether or not its origin is a plant cell. Exemplary suitable plant promoters include, but are not limited to, those that are obtained from plants, plant viruses, and bacteria such as Agrobacterium or Rhizobium which comprise genes expressed in plant cells.

[00560] As used herein, a "fungal cell" refers to any type of eukaryotic cell within the kingdom of fungi. Phyla within the kingdom of fungi include Ascomycota, Basidiomycota, Blastocladiomycota, Chytridiomycota, Glomeromycota, Microsporidia, and Neocallimastigomycota. Fungal cells may include yeasts, molds, and filamentous fungi. In some embodiments, the fungal cell is a yeast cell.

[00561] As used herein, the term "yeast cell" refers to any fungal cell within the phyla Ascomycota and Basidiomycota. Yeast cells may include budding yeast cells, fission yeast cells, and mold cells. Without being limited to these organisms, many types of yeast used in laboratory and industrial settings are part of the phylum Ascomycota. In some embodiments, the yeast cell is an S. cerervisiae, Kluyveromyces marxianus, or Issatchenkia orientalis cell. Other yeast cells may include without limitation Candida spp. (e.g., Candida albicans), Yarrowia spp. (e.g., Yarrowia lipolytica), Pichia spp. (e.g., Pichia pastoris), Kluyveromyces spp. (e.g., Kluyveromyces lactis and Kluyveromyces marxianus), Neurospora spp. (e.g., Neurospora crassa), Fusarium spp. (e.g., Fusarium oxysporum), and Issatchenkia spp. (e.g., Issatchenkia orientalis, a.k.a. Pichia kudriavzevii and Candida acidothermophilum). In some embodiments, the fungal cell is a filamentous fungal cell. As used herein, the term "filamentous fungal cell" refers to any type of fungal cell that grows in filaments, i.e., hyphae or mycelia. Examples of filamentous fungal cells may include without limitation Aspergillus spp. (e.g., Aspergillus niger), Trichoderma spp. (e.g., Trichoderma reesei), Rhizopus spp. (e.g., Rhizopus oryzae), and Mortierella spp. (e.g., Mortierella isabellina).

[00562] In some embodiments, the fungal cell is an industrial strain. As used herein, "industrial strain" refers to any strain of fungal cell used in or isolated from an industrial process, e.g., production of a product on a commercial or industrial scale. Industrial strain may refer to a fungal species that is typically used in an industrial process, or it may refer to an isolate of a fungal species that may be also used for non-industrial purposes (e.g.,

laboratory research). Examples of industrial processes may include fermentation (e.g., in production of food or beverage products), distillation, biofuel production, production of a compound, and production of a polypeptide. Examples of industrial strains may include, without limitation, JAY270 and ATCC4124.

[00563] In some embodiments, the fungal cell is a polyploid cell. As used herein, a "polyploid" cell may refer to any cell whose genome is present in more than one copy. A polyploid cell may refer to a type of cell that is naturally found in a polyploid state, or it may refer to a cell that has been induced to exist in a polyploid state (e.g., through specific regulation, alteration, inactivation, activation, or modification of meiosis, cytokinesis, or DNA replication). A polyploid cell may refer to a cell whose entire genome is polyploid, or it may refer to a cell that is polyploid in a particular genomic locus of interest. Without wishing to be bound to theory, it is thought that the abundance of guideRNA may more often be a rate-limiting component in genome engineering of polyploid cells than in haploid cells, and thus the methods using the AD-functionalized CRISPR system described herein may take advantage of using a certain fungal cell type.

[00564] In some embodiments, the fungal cell is a diploid cell. As used herein, a "diploid" cell may refer to any cell whose genome is present in two copies. A diploid cell may refer to a type of cell that is naturally found in a diploid state, or it may refer to a cell that has been induced to exist in a diploid state (e.g., through specific regulation, alteration, inactivation, activation, or modification of meiosis, cytokinesis, or DNA replication). For example, the S. cerevisiae strain S228C may be maintained in a haploid or diploid state. A diploid cell may refer to a cell whose entire genome is diploid, or it may refer to a cell that is diploid in a particular genomic locus of interest. In some embodiments, the fungal cell is a haploid cell. As used herein, a "haploid" cell may refer to any cell whose genome is present in one copy. A haploid cell may refer to a type of cell that is naturally found in a haploid state, or it may refer to a cell that has been induced to exist in a haploid state (e.g., through specific regulation, alteration, inactivation, activation, or modification of meiosis, cytokinesis, or DNA replication). For example, the S. cerevisiae strain S228C may be maintained in a haploid or diploid state. A haploid cell may refer to a cell whose entire genome is haploid, or it may refer to a cell that is haploid in a particular genomic locus of interest.

[00565] As used herein, a "yeast expression vector" refers to a nucleic acid that contains one or more sequences encoding an RNA and/or polypeptide and may further contain any desired elements that control the expression of the nucleic acid(s), as well as any elements

that enable the replication and maintenance of the expression vector inside the yeast cell. Many suitable yeast expression vectors and features thereof are known in the art; for example, various vectors and techniques are illustrated in in Yeast Protocols, 2nd edition, Xiao, W., ed. (Humana Press, New York, 2007) and Buckholz, R.G. and Gleeson, M.A. (1991) Biotechnology (NY) 9(11): 1067-72. Yeast vectors may contain, without limitation, a centromeric (CEN) sequence, an autonomous replication sequence (ARS), a promoter, such as an RNA Polymerase III promoter, operably linked to a sequence or gene of interest, a terminator such as an RNA polymerase III terminator, an origin of replication, and a marker gene (e.g., auxotrophic, antibiotic, or other selectable markers). Examples of expression vectors for use in yeast may include plasmids, yeast artificial chromosomes, 2μ plasmids, yeast integrative plasmids, yeast replicative plasmids, shuttle vectors, and episomal plasmids.

[00566] Stable integration of AD-functionalized CRISPR system components in the genome of plants and plant cells

[00567] In particular embodiments, it is envisaged that the polynucleotides encoding the components of the AD-functionalized CRISPR system are introduced for stable integration into the genome of a plant cell. In these embodiments, the design of the transformation vector or the expression system can be adjusted depending on for when, where and under what conditions the guide RNA and/or fusion protein of adenosine deaminase and Cpf1 are expressed.

[00568] In particular embodiments, it is envisaged to introduce the components of the AD-functionalized CRISPR system stably into the genomic DNA of a plant cell. Additionally or alternatively, it is envisaged to introduce the components of the AD-functionalized CRISPR system for stable integration into the DNA of a plant organelle such as, but not limited to a plastid, e mitochondrion or a chloroplast.

[00569] The expression system for stable integration into the genome of a plant cell may contain one or more of the following elements: a promoter element that can be used to express the RNA and/or fusion protein of adenosine deaminase and Cpf1 in a plant cell; a 5' untranslated region to enhance expression ; an intron element to further enhance expression in certain cells, such as monocot cells; a multiple-cloning site to provide convenient restriction sites for inserting the guide RNA and/or the fusion protein of adenosine deaminase and Cpf1 encoding sequences and other desired elements; and a 3' untranslated region to provide for efficient termination of the expressed transcript.

[00570] The elements of the expression system may be on one or more expression constructs which are either circular such as a plasmid or transformation vector, or non-circular such as linear double stranded DNA.

[00571] In a particular embodiment, a AD-functionalized CRISPR expression system comprises at least: a nucleotide sequence encoding a guide RNA (gRNA) that hybridizes with a target sequence in a plant, and wherein the guide RNA comprises a guide sequence and a direct repeat sequence, and a nucleotide sequence encoding a fusion protein of adenosine deaminase and Cpf1, wherein components (a) or (b) are located on the same or on different constructs, and whereby the different nucleotide sequences can be under control of the same or a different regulatory element operable in a plant cell.

[00572] DNA construct(s) containing the components of the AD-functionalized CRISPR system, and, where applicable, template sequence may be introduced into the genome of a plant, plant part, or plant cell by a variety of conventional techniques. The process generally comprises the steps of selecting a suitable host cell or host tissue, introducing the construct(s) into the host cell or host tissue, and regenerating plant cells or plants therefrom.

[00573] In particular embodiments, the DNA construct may be introduced into the plant cell using techniques such as but not limited to electroporation, microinjection, aerosol beam injection of plant cell protoplasts, or the DNA constructs can be introduced directly to plant tissue using biolistic methods, such as DNA particle bombardment (see also Fu et al., Transgenic Res. 2000 Feb;9(1):11-9). The basis of particle bombardment is the acceleration of particles coated with gene/s of interest toward cells, resulting in the penetration of the protoplasm by the particles and typically stable integration into the genome. (see e.g. Klein et al, Nature (1987), Klein et ah, Bio/Technology (1992), Casas et ah, Proc. Natl. Acad. Sci. USA (1993).).

[00574] In particular embodiments, the DNA constructs containing components of the AD-functionalized CRISPR system may be introduced into the plant by *Agrobacterium*-mediated transformation. The DNA constructs may be combined with suitable T-DNA flanking regions and introduced into a conventional *Agrobacterium tumefaciens* host vector. The foreign DNA can be incorporated into the genome of plants by infecting the plants or by incubating plant protoplasts with Agrobacterium bacteria, containing one or more Ti (tumor-inducing) plasmids. (see e.g. Fraley et al., (1985), Rogers et al., (1987) and U.S. Pat. No. 5,563,055).

[00575] **Plant promoters**

[00576] In order to ensure appropriate expression in a plant cell, the components of the AD-functionalized CRISPR system described herein are typically placed under control of a plant promoter, i.e. a promoter operable in plant cells. The use of different types of promoters is envisaged.

[00577] A constitutive plant promoter is a promoter that is able to express the open reading frame (ORF) that it controls in all or nearly all of the plant tissues during all or nearly all developmental stages of the plant (referred to as "constitutive expression"). One non-limiting example of a constitutive promoter is the cauliflower mosaic virus 35S promoter. "Regulated promoter" refers to promoters that direct gene expression not constitutively, but in a temporally- and/or spatially-regulated manner, and includes tissue-specific, tissue-preferred and inducible promoters. Different promoters may direct the expression of a gene in different tissues or cell types, or at different stages of development, or in response to different environmental conditions. In particular embodiments, one or more of the AD-functionalized CRISPR components are expressed under the control of a constitutive promoter, such as the cauliflower mosaic virus 35S promoter issue-preferred promoters can be utilized to target enhanced expression in certain cell types within a particular plant tissue, for instance vascular cells in leaves or roots or in specific cells of the seed. Examples of particular promoters for use in the AD-functionalized CRISPR system are found in Kawamata et al., (1997) Plant Cell Physiol 38:792-803; Yamamoto et al., (1997) Plant J 12:255-65; Hire et al, (1992) Plant Mol Biol 20:207-18,Kuster et al, (1995) Plant Mol Biol 29:759-72, and Capana et al., (1994) Plant Mol Biol 25:681 -91.

[00578] Inducible promoters can be of interest to express one or more of the components of the AD-functionalized CRISPR system under limited circumstances to avoid non-specific activity of the deaminase. In particular embodiments, one or more elements of the AD-functionalized CRISPR system are expressed under control of an inducible promoter. Examples of promoters that are inducible and that allow for spatiotemporal control of gene editing or gene expression may use a form of energy. The form of energy may include but is not limited to sound energy, electromagnetic radiation, chemical energy and/or thermal energy. Examples of inducible systems include tetracycline inducible promoters (Tet-On or Tet-Off), small molecule two-hybrid transcription activations systems (FKBP, ABA, etc), or light inducible systems (Phytochrome, LOV domains, or cryptochrome)., such as a Light Inducible Transcriptional Effector (LITE) that direct changes in transcriptional activity in a sequence-specific manner. The components of a light inducible system may include a fusion protein of adenosine deaminase and Cpf1, a light-responsive cytochrome heterodimer (e.g.

from Arabidopsis thaliana). Further examples of inducible DNA binding proteins and methods for their use are provided in US 61/736465 and US 61/721,283, which is hereby incorporated by reference in its entirety.

[00579] In particular embodiments, transient or inducible expression can be achieved by using, for example, chemical-regulated promotors, i.e. whereby the application of an exogenous chemical induces gene expression. Modulating of gene expression can also be obtained by a chemical-repressible promoter, where application of the chemical represses gene expression. Chemical-inducible promoters include, but are not limited to, the maize ln2-2 promoter, activated by benzene sulfonamide herbicide safeners (De Veylder et al., (1997) Plant Cell Physiol 38:568-77), the maize GST promoter (GST-ll-27, WO93/01294), activated by hydrophobic electrophilic compounds used as pre-emergent herbicides, and the tobacco PR-1 a promoter (Ono et al., (2004) Biosci Biotechnol Biochem 68:803-7) activated by salicylic acid. Promoters which are regulated by antibiotics, such as tetracycline-inducible and tetracycline-repressible promoters (Gatz et al., (1991) Mol Gen Genet 227:229-37; U.S. Patent Nos. 5,814,618 and 5,789,156) can also be used herein.

[00580] Translocation to and/or expression in specific plant organelles

[00581] The expression system may comprise elements for translocation to and/or expression in a specific plant organelle.

[00582] Chloroplast targeting

[00583] In particular embodiments, it is envisaged that the AD-functionalized CRISPR system is used to specifically modify chloroplast genes or to ensure expression in the chloroplast. For this purpose use is made of chloroplast transformation methods or compartimentalization of the AD-functionalized CRISPR components to the chloroplast. For instance, the introduction of genetic modifications in the plastid genome can reduce biosafety issues such as gene flow through pollen.

[00584] Methods of chloroplast transformation are known in the art and include Particle bombardment, PEG treatment, and microinjection. Additionally, methods involving the translocation of transformation cassettes from the nuclear genome to the pastid can be used as described in WO2010061186.

[00585] Alternatively, it is envisaged to target one or more of the AD-functionalized CRISPR components to the plant chloroplast. This is achieved by incorporating in the expression construct a sequence encoding a chloroplast transit peptide (CTP) or plastid transit peptide, operably linked to the 5' region of the sequence encoding the fusion protein of adenosine deaminase and Cpf1. The CTP is removed in a processing step during

translocation into the chloroplast. Chloroplast targeting of expressed proteins is well known to the skilled artisan (see for instance Protein Transport into Chloroplasts, 2010, Annual Review of Plant Biology,Vol. 61: 157-180) . In such embodiments it is also desired to target the guide RNA to the plant chloroplast. Methods and constructs which can be used for translocating guide RNA into the chloroplast by means of a chloroplast localization sequence are described, for instance, in US 20040142476, incorporated herein by reference. Such variations of constructs can be incorporated into the expression systems of the invention to efficiently translocate the AD-functionalized CRISPR system components.

[00586] Introduction of polynucleotides encoding the AD-functionalized CRISPR system in Algae cells.

[00587] Transgenic algae (or other plants such as rape) may be particularly useful in the production of vegetable oils or biofuels such as alcohols (especially methanol and ethanol) or other products. These may be engineered to express or overexpress high levels of oil or alcohols for use in the oil or biofuel industries.

[00588] US 8945839 describes a method for engineering Micro-Algae (*Chlamydomonas reinhardtii* cells) species) using Cas9. Using similar tools, the methods of the AD-functionalized CRISPR system described herein can be applied on *Chlamydomonas* species and other algae. In particular embodiments, a CRISPR-Cas protein (e.g., Cpf1), adenosine deaminase (which may be fused to the CRISPR-Cas protein or an aptamer-binding adaptor protein), and guide RNA are introduced in algae expressed using a vector that expresses the fusion protein of adenosine deaminase and Cpf1 under the control of a constitutive promoter such as Hsp70A-Rbc S2 or Beta2 -tubulin. Guide RNA is optionally delivered using a vector containing T7 promoter. Alternatively, Cpf1 mRNA and in vitro transcribed guide RNA can be delivered to algal cells. Electroporation protocols are available to the skilled person such as the standard recommended protocol from the GeneArt Chlamydomonas Engineering kit.

[00589] Introduction of AD-functionalized CRISPR system components in yeast cells

[00590] In particular embodiments, the invention relates to the use of the AD-functionalized CRISPR system for genome editing of yeast cells. Methods for transforming yeast cells which can be used to introduce polynucleotides encoding the AD-functionalized CRISPR system components are described in Kawai et al., 2010, Bioeng Bugs. 2010 Nov-Dec; 1(6): 395–403). Non-limiting examples include transformation of yeast cells by

lithium acetate treatment (which may further include carrier DNA and PEG treatment), bombardment or by electroporation.

[00591] Transient expression of AD-functionalized CRISPR system components in plants and plant cell

[00592] In particular embodiments, it is envisaged that the guide RNA and/or CRISPR-Cas gene are transiently expressed in the plant cell. In these embodiments, the AD-functionalized CRISPR system can ensure modification of a target gene only when both the guide RNA, the CRISPR-Cas protein (e.g., Cpf1), and adenosine deaminase (which may be fused to the CRISPR-Cas protein or an aptamer-binding adaptor protein), are present in a cell, such that genomic modification can further be controlled. As the expression of the CRISPR-Cas protein is transient, plants regenerated from such plant cells typically contain no foreign DNA. In particular embodiments the CRISPR-Cas protein is stably expressed by the plant cell and the guide sequence is transiently expressed.

[00593] In particular embodiments, the AD-functionalized CRISPR system components can be introduced in the plant cells using a plant viral vector (Scholthof et al. 1996, Annu Rev Phytopathol. 1996;34:299-323). In further particular embodiments, said viral vector is a vector from a DNA virus. For example, geminivirus (e.g., cabbage leaf curl virus, bean yellow dwarf virus, wheat dwarf virus, tomato leaf curl virus, maize streak virus, tobacco leaf curl virus, or tomato golden mosaic virus) or nanovirus (e.g., Faba bean necrotic yellow virus). In other particular embodiments, said viral vector is a vector from an RNA virus. For example, tobravirus (e.g., tobacco rattle virus, tobacco mosaic virus), potexvirus (e.g., potato virus X), or hordeivirus (e.g., barley stripe mosaic virus). The replicating genomes of plant viruses are non-integrative vectors.

[00594] In particular embodiments, the vector used for transient expression of AD-functionalized CRISPR system is for instance a pEAQ vector, which is tailored for Agrobacterium-mediated transient expression (Sainsbury F. et al., Plant Biotechnol J. 2009 Sep;7(7):682-93) in the protoplast. Precise targeting of genomic locations was demonstrated using a modified Cabbage Leaf Curl virus (CaLCuV) vector to express guide RNAs in stable transgenic plants expressing a CRISPR enzyme (Scientific Reports 5, Article number: 14926 (2015), doi:10.1038/srep14926).

[00595] In particular embodiments, double-stranded DNA fragments encoding the guide RNA and/or the CRISPR-Cas gene can be transiently introduced into the plant cell. In such embodiments, the introduced double-stranded DNA fragments are provided in sufficient quantity to modify the cell but do not persist after a contemplated period of time has passed

or after one or more cell divisions. Methods for direct DNA transfer in plants are known by the skilled artisan (see for instance Davey et al. Plant Mol Biol. 1989 Sep;13(3):273-85.)

[00596] In other embodiments, an RNA polynucleotide encoding the CRISPR-Cas protein (e.g., Cpf1) and/or adenosine deaminase (which may be fused to the CRISPR-Cas protein or an aptamer-binding adaptor protein) is introduced into the plant cell, which is then translated and processed by the host cell generating the protein in sufficient quantity to modify the cell (in the presence of at least one guide RNA) but which does not persist after a contemplated period of time has passed or after one or more cell divisions. Methods for introducing mRNA to plant protoplasts for transient expression are known by the skilled artisan (see for instance in Gallie, Plant Cell Reports (1993), 13;119-122).

[00597] Combinations of the different methods described above are also envisaged.

[00598] **Delivery of AD-functionalized CRISPR system components to the plant cell**

[00599] In particular embodiments, it is of interest to deliver one or more components of the AD-functionalized CRISPR system directly to the plant cell. This is of interest, inter alia, for the generation of non-transgenic plants (see below). In particular embodiments, one or more of the AD-functionalized CRISPR system components is prepared outside the plant or plant cell and delivered to the cell. For instance in particular embodiments, the CRISPR-Cas protein is prepared in vitro prior to introduction to the plant cell. The CRISPR-Cas protein can be prepared by various methods known by one of skill in the art and include recombinant production. After expression, the CRISPR-Cas protein is isolated, refolded if needed, purified and optionally treated to remove any purification tags, such as a His-tag. Once crude, partially purified, or more completely purified CRISPR-Cas protein is obtained, the protein may be introduced to the plant cell.

[00600] In particular embodiments, the CRISPR-Cas protein is mixed with guide RNA targeting the gene of interest to form a pre-assembled ribonucleoprotein.

[00601] The individual components or pre-assembled ribonucleoprotein can be introduced into the plant cell via electroporation, by bombardment with CRISPR-Cas-associated gene product coated particles, by chemical transfection or by some other means of transport across a cell membrane. For instance, transfection of a plant protoplast with a pre-assembled CRISPR ribonucleoprotein has been demonstrated to ensure targeted modification of the plant genome (as described by Woo et al. *Nature Biotechnology*, 2015; DOI: 10.1038/nbt.3389).

[00602] In particular embodiments, the AD-functionalized CRISPR system components are introduced into the plant cells using nanoparticles. The components, either as protein or

nucleic acid or in a combination thereof, can be uploaded onto or packaged in nanoparticles and applied to the plants (such as for instance described in WO 2008042156 and US 20130185823). In particular, embodiments of the invention comprise nanoparticles uploaded with or packed with DNA molecule(s) encoding the CRISPR-Cas protein (e.g., Cpf1), DNA molecule(s) encoding adenosine deaminase (which may be fused to the CRISPR-Cas protein or an aptamer-binding adaptor protein), and DNA molecules encoding the guide RNA and/or isolated guide RNA as described in WO2015089419.

[00603] Further means of introducing one or more components of the AD-functionalized CRISPR system to the plant cell is by using cell penetrating peptides (CPP). Accordingly, in particular, embodiments the invention comprises compositions comprising a cell penetrating peptide linked to the CRISPR-Cas protein. In particular embodiments of the present invention, the CRISPR-Cas protein and/or guide RNA is coupled to one or more CPPs to effectively transport them inside plant protoplasts. Ramakrishna (Genome Res. 2014 Jun;24(6):1020-7 for Cas9 in human cells). In other embodiments, the CRISPR-Cas gene and/or guide RNA are encoded by one or more circular or non-circular DNA molecule(s) which are coupled to one or more CPPs for plant protoplast delivery. The plant protoplasts are then regenerated to plant cells and further to plants. CPPs are generally described as short peptides of fewer than 35 amino acids either derived from proteins or from chimeric sequences which are capable of transporting biomolecules across cell membrane in a receptor independent manner. CPP can be cationic peptides, peptides having hydrophobic sequences, amphipatic peptides, peptides having proline-rich and anti-microbial sequence, and chimeric or bipartite peptides (Pooga and Langel 2005). CPPs are able to penetrate biological membranes and as such trigger the movement of various biomolecules across cell membranes into the cytoplasm and to improve their intracellular routing, and hence facilitate interaction of the biolomolecule with the target. Examples of CPP include amongst others: Tat, a nuclear transcriptional activator protein required for viral replication by HIV type1, penetratin, Kaposi fibroblast growth factor (FGF) signal peptide sequence, integrin β3 signal peptide sequence; polyarginine peptide Args sequence, Guanine rich-molecular transporters, sweet arrow peptide, etc.

[00604] Use of the AD-functionalized CRISPR system to make genetically modified non-transgenic plants

[00605] In particular embodiments, the methods described herein are used to modify endogenous genes or to modify their expression without the permanent introduction into the genome of the plant of any foreign gene, including those encoding CRISPR components, so

as to avoid the presence of foreign DNA in the genome of the plant. This can be of interest as the regulatory requirements for non-transgenic plants are less rigorous.

[00606] In particular embodiments, this is ensured by transient expression of the AD-functionalized CRISPR system components . In particular embodiments one or more of the components are expressed on one or more viral vectors which produce sufficient CRISPR-Cas protein, adenosine deaminase, and guide RNA to consistently steadily ensure modification of a gene of interest according to a method described herein.

[00607] In particular embodiments, transient expression of AD-functionalized CRISPR system constructs is ensured in plant protoplasts and thus not integrated into the genome. The limited window of expression can be sufficient to allow the AD-functionalized CRISPR system to ensure modification of a target gene as described herein.

[00608] In particular embodiments, the different components of the AD-functionalized CRISPR system are introduced in the plant cell, protoplast or plant tissue either separately or in mixture, with the aid of pariculate delivering molecules such as nanoparticles or CPP molecules as described herein above.

[00609] The expression of the AD-functionalized CRISPR system components can induce targeted modification of the genome, by deaminase activity of the adenosine deaminase. The different strategies described herein above allow CRISPR-mediated targeted genome editing without requiring the introduction of the AD-functionalized CRISPR systemt components into the plant genome. Components which are transiently introduced into the plant cell are typically removed upon crossing.

[00610] Plant cultures and regeneration

[00611] In particular embodiments, plant cells which have a modified genome and that are produced or obtained by any of the methods described herein, can be cultured to regenerate a whole plant which possesses the transformed or modified genotype and thus the desired phenotype. Conventional regeneration techniques are well known to those skilled in the art. Particular examples of such regeneration techniques rely on manipulation of certain phytohormones in a tissue culture growth medium, and typically relying on a biocide and/or herbicide marker which has been introduced together with the desired nucleotide sequences. In further particular embodiments, plant regeneration is obtained from cultured protoplasts, plant callus, explants, organs, pollens, embryos or parts thereof (see e.g. Evans et al. (1983), Handbook of Plant Cell Culture, Klee et al (1987) Ann. Rev. of Plant Phys.).

[00612] In particular embodiments, transformed or improved plants as described herein can be self-pollinated to provide seed for homozygous improved plants of the invention (homozygous for the DNA modification) or crossed with non-transgenic plants or different improved plants to provide seed for heterozygous plants. Where a recombinant DNA was introduced into the plant cell, the resulting plant of such a crossing is a plant which is heterozygous for the recombinant DNA molecule. Both such homozygous and heterozygous plants obtained by crossing from the improved plants and comprising the genetic modification (which can be a recombinant DNA) are referred to herein as "progeny". Progeny plants are plants descended from the original transgenic plant and containing the genome modification or recombinant DNA molecule introduced by the methods provided herein. Alternatively, genetically modified plants can be obtained by one of the methods described supra using the AD-functionalized CRISPR system whereby no foreign DNA is incorporated into the genome. Progeny of such plants, obtained by further breeding may also contain the genetic modification. Breedings are performed by any breeding methods that are commonly used for different crops (e.g., Allard, Principles of Plant Breeding, John Wiley & Sons, NY, U. of CA, Davis, CA, 50-98 (1960).

[00613] Generation of plants with enhanced agronomic traits

[00614] The AD-functionalized CRISPR systems provided herein can be used to introduce targeted A-G and T-C mutations. By co-expression of multiple targeting RNAs directed to achieve multiple modifications in a single cell, multiplexed genome modification can be ensured. This technology can be used to high-precision engineering of plants with improved characteristics, including enhanced nutritional quality, increased resistance to diseases and resistance to biotic and abiotic stress, and increased production of commercially valuable plant products or heterologous compounds.

[00615] In particular embodiments, the AD-functionalized CRISPR system as described herein is used to introduce targeted A-G and T-C mutations. Such mutation can be a nonsense mutation (e.g., premature stop codon) or a missense mutation (e.g., encoding different amino acid residue). This is of interest where the A-G and T-C mutations in certain endogenous genes can confer or contribute to a desired trait.

[00616] The methods described herein generally result in the generation of "improved plants" in that they have one or more desirable traits compared to the wildtype plant. In particular embodiments, the plants, plant cells or plant parts obtained are transgenic plants, comprising an exogenous DNA sequence incorporated into the genome of all or part of the cells of the plant. In particular embodiments, non-transgenic genetically modified plants,

plant parts or cells are obtained, in that no exogenous DNA sequence is incorporated into the genome of any of the plant cells of the plant. In such embodiments, the improved plants are non-transgenic. Where only the modification of an endogenous gene is ensured and no foreign genes are introduced or maintained in the plant genome, the resulting genetically modified crops contain no foreign genes and can thus basically be considered non-transgenic.

[00617] In particular embodiments, the polynucleotides are delivered into the cell by a DNA virus (e.g., a geminivirus) or an RNA virus (e.g., a tobravirus). In particular embodiments, the introducing steps include delivering to the plant cell a T-DNA containing one or more polynucleotide sequences encoding the CRISPR-Cas protein, the adenosine deaminase, and the guide RNA, where the delivering is via Agrobacterium. The polynucleotide sequence encoding the components of the AD-functionalized CRISPR system can be operably linked to a promoter, such as a constitutive promoter (e.g., a cauliflower mosaic virus 35S promoter), or a cell specific or inducible promoter. In particular embodiments, the polynucleotide is introduced by microprojectile bombardment. In particular embodiments, the method further includes screening the plant cell after the introducing steps to determine whether the expression of the gene of interest has been modified. In particular embodiments, the methods include the step of regenerating a plant from the plant cell. In further embodiments, the methods include cross breeding the plant to obtain a genetically desired plant lineage.

[00618] In particular embodiments of the methods described above, disease resistant crops are obtained by targeted mutation of disease susceptibility genes or genes encoding negative regulators (e.g. Mlo gene) of plant defense genes. In a particular embodiment, herbicide-tolerant crops are generated by targeted substitution of specific nucleotides in plant genes such as those encoding acetolactate synthase (ALS) and protoporphyrinogen oxidase (PPO). In particular embodiments drought and salt tolerant crops by targeted mutation of genes encoding negative regulators of abiotic stress tolerance, low amylose grains by targeted mutation of Waxy gene, rice or other grains with reduced rancidity by targeted mutation of major lipase genes in aleurone layer, etc. In particular embodiments. A more extensive list of endogenous genes encoding a traits of interest are listed below.

[00619] Use of AD-functionalized CRISPR system to modify polyploid plants

[00620] Many plants are polyploid, which means they carry duplicate copies of their genomes—sometimes as many as six, as in wheat. The methods according to the present invention, which make use of the AD-functionalized CRISPR system can be “multiplexed”

to affect all copies of a gene, or to target dozens of genes at once. For instance, in particular embodiments, the methods of the present invention are used to simultaneously ensure a loss of function mutation in different genes responsible for suppressing defences against a disease. In particular embodiments, the methods of the present invention are used to simultaneously suppress the expression of the TaMLO-Al, TaMLO-Bl and TaMLO-Dl nucleic acid sequence in a wheat plant cell and regenerating a wheat plant therefrom, in order to ensure that the wheat plant is resistant to powdery mildew (see also WO2015109752).

[00621] Examplary genes conferring agronomic traits

[00622] In particular embodiments, the invention encompasses methods which involve targeted A-G and T-C mutations in endogenous genes and their regulatory elements, such as listed below:

[00623] 1. Genes that confer resistance to pests or diseases:

[00624] Plant disease resistance genes. A plant can be transformed with cloned resistance genes to engineer plants that are resistant to specific pathogen strains. See, e.g., Jones et al., Science 266:789 (1994) (cloning of the tomato Cf- 9 gene for resistance to Cladosporium fulvum); Martin et al., Science 262:1432 (1993) (tomato Pto gene for resistance to Pseudomonas syringae pv. tomato encodes a protein kinase); Mindrinos et al., Cell 78:1089 (1994) (Arabidopsmay be RSP2 gene for resistance to Pseudomonas syringae). A plant gene that is upregulated or down regulated during pathogen infection can be engineered for pathogen resistance. See, e.g., Thomazella et al., bioRxiv 064824; doi: https://doi.org/10.1101/064824 Epub. July 23, 2016 (tomato plants with deletions in the SlDMR6-1 which is normally upregulated during pathogen infection).

[00625] Genes conferring resistance to a pest, such as soybean cyst nematode. See e.g., PCT Application WO 96/30517; PCT Application WO 93/19181.

[00626] Bacillus thuringiensis proteins see, e.g., Geiser et al., Gene 48:109 (1986).

[00627] Lectins, see, for example, Van Damme et al., Plant Molec. Biol. 24:25 (1994.

[00628] Vitamin-binding protein, such as avidin, see PCT application US93/06487, teaching the use of avidin and avidin homologues as larvicides against insect pests.

[00629] Enzyme inhibitors such as protease or proteinase inhibitors or amylase inhibitors. See, e.g., Abe et al., J. Biol. Chem. 262:16793 (1987), Huub et al., Plant Molec. Biol. 21:985 (1993)), Sumitani et al., Biosci. Biotech. Biochem. 57:1243 (1993) and U.S. Pat. No. 5,494,813.

[00630] Insect-specific hormones or pheromones such as ecdysteroid or juvenile hormone, a variant thereof, a mimetic based thereon, or an antagonist or agonist thereof. See, for example Hammock et al., Nature 344:458 (1990).

[00631] Insect-specific peptides or neuropeptides which, upon expression, disrupts the physiology of the affected pest. For example Regan, J. Biol. Chem. 269:9 (1994) and Pratt et al., Biochem. Biophys. Res. Comm. 163:1243 (1989). See also U.S. Pat. No. 5,266,317.

[00632] Insect-specific venom produced in nature by a snake, a wasp, or any other organism. For example, see Pang et al., Gene 116: 165 (1992).

[00633] Enzymes responsible for a hyperaccumulation of a monoterpene, a sesquiterpene, a steroid, hydroxamic acid, a phenylpropanoid derivative or another nonprotein molecule with insecticidal activity.

[00634] Enzymes involved in the modification, including the post-translational modification, of a biologically active molecule; for example, a glycolytic enzyme, a proteolytic enzyme, a lipolytic enzyme, a nuclease, a cyclase, a transaminase, an esterase, a hydrolase, a phosphatase, a kinase, a phosphorylase, a polymerase, an elastase, a chitinase and a glucanase, whether natural or synthetic. See PCT application WO93/02197, Kramer et al., Insect Biochem. Molec. Biol. 23:691 (1993) and Kawalleck et al., Plant Molec. Biol. 21 :673 (1993).

[00635] Molecules that stimulates signal transduction. For example, see Botella et al., Plant Molec. Biol. 24:757 (1994), and Griess et al., Plant Physiol. 104:1467 (1994).

[00636] Viral-invasive proteins or a complex toxin derived therefrom.See Beachy et al., Ann. rev. Phytopathol. 28:451 (1990).

[00637] Developmental-arrestive proteins produced in nature by a pathogen or a parasite. See Lamb et al., Bio/Technology 10:1436 (1992) and Toubart et al., Plant J. 2:367 (1992).

[00638] A developmental-arrestive protein produced in nature by a plant. For example, Logemann et al., Bio/Technology 10:305 (1992).

[00639] In plants, pathogens are often host-specific. For example, some Fusarium species will causes tomato wilt but attacks only tomato, and other Fusarium species attack only wheat. Plants have existing and induced defenses to resist most pathogens. Mutations and recombination events across plant generations lead to genetic variability that gives rise to susceptibility, especially as pathogens reproduce with more frequency than plants. In plants there can be non-host resistance, e.g., the host and pathogen are incompatible or there can be partial resistance against all races of a pathogen, typically controlled by many genes and/or also complete resistance to some races of a pathogen but not to other races. Such

resistance is typically controlled by a few genes. Using methods and components of the AD-functionalized CRISPR system, a new tool now exists to induce specific mutations in anticipation hereon. Accordingly, one can analyze the genome of sources of resistance genes, and in plants having desired characteristics or traits, use the method and components of the AD-functionalized CRISPR system to induce the rise of resistance genes. The present systems can do so with more precision than previous mutagenic agents and hence accelerate and improve plant breeding programs.

[00640] 2. Genes involved in plant diseases, such as those listed in WO 2013046247:

[00641] Rice diseases: Magnaporthe grisea, Cochliobolus miyabeanus, Rhizoctonia solani, Gibberella fujikuroi; Wheat diseases: Erysiphe graminis, Fusarium graminearum, F. avenaceum, F. culmorum, Microdochium nivale, Puccinia striiformis, P. graminis, P. recondita, Micronectriella nivale, Typhula sp., Ustilago tritici, Tilletia caries, Pseudocercosporella herpotrichoides, Mycosphaerella graminicola, Stagonospora nodorum, Pyrenophora tritici-repentis;Barley diseases: Erysiphe graminis, Fusarium graminearum, F. avenaceum, F. culmorum, Microdochium nivale, Puccinia striiformis, P. graminis, P. hordei, Ustilago nuda, Rhynchosporium secalis, Pyrenophora teres, Cochliobolus sativus, Pyrenophora graminea, Rhizoctonia solani;Maize diseases: Ustilago maydis, Cochliobolus heterostrophus, Gloeocercospora sorghi, Puccinia polysora, Cercospora zeae-maydis, Rhizoctonia solani;

[00642] Citrus diseases: Diaporthe citri, Elsinoe fawcetti, Penicillium digitatum, P. italicum, Phytophthora parasitica, Phytophthora citrophthora;Apple diseases: Monilinia mali, Valsa ceratosperma, Podosphaera leucotricha, Alternaria alternata apple pathotype, Venturia inaequalis, Colletotrichum acutatum, Phytophtora cactorum;

[00643] Pear diseases: Venturia nashicola, V. pirina, Alternaria alternata Japanese pear pathotype, Gymnosporangium haraeanum, Phytophtora cactorum;

[00644] Peach diseases: Monilinia fructicola, Cladosporium carpophilum, Phomopsis sp.;

[00645] Grape diseases: Elsinoe ampelina, Glomerella cingulata, Uninula necator, Phakopsora ampelopsidis, Guignardia bidwellii, Plasmopara viticola;

[00646] Persimmon diseases: Gloesporium kaki, Cercospora kaki, Mycosphaerela nawae;

[00647] Gourd diseases: Colletotrichum lagenarium, Sphaerotheca fuliginea, Mycosphaerella melonis, Fusarium oxysporum, Pseudoperonospora cubensis, Phytophthora sp., Pythium sp.;

[00648] Tomato diseases: Alternaria solani, Cladosporium fulvum, Phytophthora infestans; Pseudomonas syringae pv. Tomato; Phytophthora capsici; Xanthomonas

[00649] Eggplant diseases: Phomopsis vexans, Erysiphe cichoracearum; Brassicaceous vegetable diseases: Alternaria japonica, Cercosporella brassicae, Plasmodiophora brassicae, Peronospora parasitica;

[00650] Welsh onion diseases: Puccinia allii, Peronospora destructor;

[00651] Soybean diseases: Cercospora kikuchii, Elsinoe glycines, Diaporthe phaseolorum var. sojae, Septoria glycines, Cercospora sojina, Phakopsora pachyrhizi, Phytophthora sojae, Rhizoctonia solani, Corynespora casiicola, Sclerotinia sclerotiorum;

[00652] Kidney bean diseases: Colletrichum lindemthianum;

[00653] Peanut diseases: Cercospora personata, Cercospora arachidicola, Sclerotium rolfsii;

[00654] Pea diseases pea: Erysiphe pisi;

[00655] Potato diseases: Alternaria solani, Phytophthora infestans, Phytophthora erythroseptica, Spongospora subterranean, f. sp. Subterranean;

[00656] Strawberry diseases: Sphaerotheca humuli, Glomerella cingulata;

[00657] Tea diseases: Exobasidium reticulatum, Elsinoe leucospila, Pestalotiopsis sp., Colletotrichum theae-sinensis;

[00658] Tobacco diseases: Alternaria longipes, Erysiphe cichoracearum, Colletotrichum tabacum, Peronospora tabacina, Phytophthora nicotianae;

[00659] Rapeseed diseases: Sclerotinia sclerotiorum, Rhizoctonia solani;

[00660] Cotton diseases: Rhizoctonia solani;

[00661] Beet diseases: Cercospora beticola, Thanatephorus cucumeris, Thanatephorus cucumeris, Aphanomyces cochlioides;

[00662] Rose diseases: Diplocarpon rosae, Sphaerotheca pannosa, Peronospora sparsa;

[00663] Diseases of chrysanthemum and asteraceae: Bremia lactuca, Septoria chrysanthemi-indici, Puccinia horiana;

[00664] Diseases of various plants: Pythium aphanidermatum, Pythium debarianum, Pythium graminicola, Pythium irregulare, Pythium ultimum, Botrytis cinerea, Sclerotinia sclerotiorum;

[00665] Radish diseases: Alternaria brassicicola;

[00666] Zoysia diseases: Sclerotinia homeocarpa, Rhizoctonia solani;

[00667] Banana diseases: Mycosphaerella fijiensis, Mycosphaerella musicola;

[00668] Sunflower diseases: Plasmopara halstedii;

[00669] Seed diseases or diseases in the initial stage of growth of various plants caused by Aspergillus spp., Penicillium spp., Fusarium spp., Gibberella spp., Tricoderma spp., Thielaviopsis spp., Rhizopus spp., Mucor spp., Corticium spp., Rhoma spp., Rhizoctonia spp., Diplodia spp., or the like;

[00670] Virus diseases of various plants mediated by Polymixa spp., Olpidium spp., or the like.

[00671] 3. Examples of genes that confer resistance to herbicides:

[00672] Resistance to herbicides that inhibit the growing point or meristem, such as an imidazolinone or a sulfonylurea, for example, by Lee et al., EMBO J. 7:1241 (1988), and Miki et al., Theor. Appl. Genet. 80:449 (1990), respectively.

[00673] Glyphosate tolerance (resistance conferred by, e.g., mutant 5-enolpyruvylshikimate-3- phosphate synthase (EPSPs) genes, aroA genes and glyphosate acetyl transferase (GAT) genes, respectively), or resistance to other phosphono compounds such as by glufosinate (phosphinothricin acetyl transferase (PAT) genes from Streptomyces species, including Streptomyces hygroscopicus and Streptomyces viridichromogenes), and to pyridinoxy or phenoxy proprionic acids and cyclohexones by ACCase inhibitor-encoding genes. See, for example, U.S. Pat. No. 4,940,835 and U.S. Pat. 6,248,876 , U.S. Pat. No. 4,769,061 , EP No. 0 333 033 and U.S. Pat No. 4,975,374. See also EP No. 0242246, DeGreef et al., Bio/Technology 7:61 (1989), Marshall et al., Theor. Appl. Genet. 83:435 (1992), WO 2005012515 to Castle et. al. and WO 2005107437.

[00674] Resistance to herbicides that inhibit photosynthesis, such as a triazine (psbA and gs+ genes) or a benzonitrile (nitrilase gene), and glutathione S-transferase in Przibila et al., Plant Cell 3:169 (1991), U.S. Pat. No. 4,810,648, and Hayes et al., Biochem. J. 285: 173 (1992).

[00675] Genes encoding Enzymes detoxifying the herbicide or a mutant glutamine synthase enzyme that is resistant to inhibition, e.g. n U.S. patent application Ser. No. 11/760,602. Or a detoxifying enzyme is an enzyme encoding a phosphinothricin acetyltransferase (such as the bar or pat protein from Streptomyces species). Phosphinothricin acetyltransferases are for example described in U.S. Pat. Nos. 5,561,236; 5,648,477; 5,646,024; 5,273,894; 5,637,489; 5,276,268; 5,739,082; 5,908,810 and 7,112,665.

[00676] Hydroxyphenylpyruvatedioxygenases (HPPD) inhibitors, naturally occuring HPPD resistant enzymes, or genes encoding a mutated or chimeric HPPD enzyme as

described in WO 96/38567, WO 99/24585, and WO 99/24586, WO 2009/144079, WO 2002/046387, or U.S. Pat. No. 6,768,044.

[00677] 4. Examples of genes involved in Abiotic stress tolerance:

[00678] Transgene capable of reducing the expression and/or the activity of poly(ADP-ribose) polymerase (PARP) gene in the plant cells or plants as described in WO 00/04173 or, WO/2006/045633.

[00679] Transgenes capable of reducing the expression and/or the activity of the PARG encoding genes of the plants or plants cells, as described e.g. in WO 2004/090140.

[00680] Transgenes coding for a plant-functional enzyme of the nicotineamide adenine dinucleotide salvage synthesis pathway including nicotinamidase, nicotinate phosphoribosyltransferase, nicotinic acid mononucleotide adenyl transferase, nicotinamide adenine dinucleotide synthetase or nicotine amide phosphorybosyltransferase as described e.g. in EP 04077624.7, WO 2006/133827, PCT/EP07/002,433, EP 1999263, or WO 2007/107326.

[00681] Enzymes involved in carbohydrate biosynthesis include those described in e.g. EP 0571427, WO 95/04826, EP 0719338, WO 96/15248, WO 96/19581, WO 96/27674, WO 97/11188, WO 97/26362, WO 97/32985, WO 97/42328, WO 97/44472, WO 97/45545, WO 98/27212, WO 98/40503, WO99/58688, WO 99/58690, WO 99/58654, WO 00/08184, WO 00/08185, WO 00/08175, WO 00/28052, WO 00/77229, WO 01/12782, WO 01/12826, WO 02/101059, WO 03/071860, WO 2004/056999, WO 2005/030942, WO 2005/030941, WO 2005/095632, WO 2005/095617, WO 2005/095619, WO 2005/095618, WO 2005/123927, WO 2006/018319, WO 2006/103107, WO 2006/108702, WO 2007/009823, WO 00/22140, WO 2006/063862, WO 2006/072603, WO 02/034923, EP 06090134.5, EP 06090228.5, EP 06090227.7, EP 07090007.1, EP 07090009.7, WO 01/14569, WO 02/79410, WO 03/33540, WO 2004/078983, WO 01/19975, WO 95/26407, WO 96/34968, WO 98/20145, WO 99/12950, WO 99/66050, WO 99/53072, U.S. Pat. No. 6,734,341, WO 00/11192, WO 98/22604, WO 98/32326, WO 01/98509, WO 01/98509, WO 2005/002359, U.S. Pat. No. 5,824,790, U.S. Pat. No. 6,013,861, WO 94/04693, WO 94/09144, WO 94/11520, WO 95/35026 or WO 97/20936 or enzymes involved in the production of polyfructose, especially of the inulin and levan-type, as disclosed in EP 0663956, WO 96/01904, WO 96/21023, WO 98/39460, and WO 99/24593, the production of alpha-1,4-glucans as disclosed in WO 95/31553, US 2002031826, U.S. Pat. No. 6,284,479, U.S. Pat. No. 5,712,107, WO 97/47806, WO 97/47807, WO 97/47808 and WO 00/14249, the production of alpha-1,6 branched alpha-1,4-glucans, as disclosed in WO 00/73422, the

production of alternan, as disclosed in e.g. WO 00/47727, WO 00/73422, EP 06077301.7, U.S. Pat. No. 5,908,975 and EP 0728213, the production of hyaluronan, as for example disclosed in WO 2006/032538, WO 2007/039314, WO 2007/039315, WO 2007/039316, JP 2006304779, and WO 2005/012529.

[00682] Genes that improve drought resistance. For example, WO 2013122472 discloses that the absence or reduced level of functional Ubiquitin Protein Ligase protein (UPL) protein, more specifically, UPL3, leads to a decreased need for water or improved resistance to drought of said plant. Other examples of transgenic plants with increased drought tolerance are disclosed in, for example, US 2009/0144850, US 2007/0266453, and WO 2002/083911. US2009/0144850 describes a plant displaying a drought tolerance phenotype due to altered expression of a DR02 nucleic acid. US 2007/0266453 describes a plant displaying a drought tolerance phenotype due to altered expression of a DR03 nucleic acid and WO 2002/08391 1 describes a plant having an increased tolerance to drought stress due to a reduced activity of an ABC transporter which is expressed in guard cells. Another example is the work by Kasuga and co-authors (1999), who describe that overexpression of cDNA encoding DREB1 A in transgenic plants activated the expression of many stress tolerance genes under normal growing conditions and resulted in improved tolerance to drought, salt loading, and freezing. However, the expression of DREB1A also resulted in severe growth retardation under normal growing conditions (Kasuga (1999) Nat Biotechnol 17(3) 287-291).

[00683] In further particular embodiments, crop plants can be improved by influencing specific plant traits. For example, by developing pesticide-resistant plants, improving disease resistance in plants, improving plant insect and nematode resistance, improving plant resistance against parasitic weeds, improving plant drought tolerance, improving plant nutritional value, improving plant stress tolerance, avoiding self-pollination, plant forage digestibility biomass, grain yield etc. A few specific non-limiting examples are provided hereinbelow.

[00684] In addition to targeted mutation of single genes, AD-functionalized CRISPR system can be designed to allow targeted mutation of multiple genes, deletion of chromosomal fragment, site-specific integration of transgene, site-directed mutagenesis in vivo, and precise gene replacement or allele swapping in plants. Therefore, the methods described herein have broad applications in gene discovery and validation, mutational and cisgenic breeding, and hybrid breeding. These applications facilitate the production of a new generation of genetically modified crops with various improved agronomic traits such

as herbicide resistance, disease resistance, abiotic stress tolerance, high yield, and superior quality.

[00685] Use of AD-functionalized CRISPR system to create male sterile plants

[00686] Hybrid plants typically have advantageous agronomic traits compared to inbred plants. However, for self-pollinating plants, the generation of hybrids can be challenging. In different plant types, genes have been identified which are important for plant fertility, more particularly male fertility. For instance, in maize, at least two genes have been identified which are important in fertility (Amitabh Mohanty International Conference on New Plant Breeding Molecular Technologies Technology Development And Regulation, Oct 9-10, 2014, Jaipur, India; Svitashev et al. Plant Physiol. 2015 Oct;169(2):931-45; Djukanovic et al. Plant J. 2013 Dec;76(5):888-99). The methods and systems provided herein can be used to target genes required for male fertility so as to generate male sterile plants which can easily be crossed to generate hybrids. In particular embodiments, the AD-functionalized CRISPR system provided herein is used for targeted mutagenesis of the cytochrome P450-like gene (MS26) or the meganuclease gene (MS45) thereby conferring male sterility to the maize plant. Maize plants which are as such genetically altered can be used in hybrid breeding programs.

[00687] Increasing the fertility stage in plants

[00688] In particular embodiments, the methods and systems provided herein are used to prolong the fertility stage of a plant such as of a rice plant. For instance, a rice fertility stage gene such as Ehd3 can be targeted in order to generate a mutation in the gene and plantlets can be selected for a prolonged regeneration plant fertility stage (as described in CN 104004782)

[00689] Use of AD-functionalized CRISPR system to generate genetic variation in a crop of interest

[00690] The availability of wild germplasm and genetic variations in crop plants is the key to crop improvement programs, but the available diversity in germplasms from crop plants is limited. The present invention envisages methods for generating a diversity of genetic variations in a germplasm of interest. In this application of the AD-functionalized CRISPR system a library of guide RNAs targeting different locations in the plant genome is provided and is introduced into plant cells together with the CRISPR-Cas protein and adenosine deaminase. In this way a collection of genome-scale point mutations and gene knock-outs can be generated. In particular embodiments, the methods comprise generating a plant part or plant from the cells so obtained and screening the cells for a trait of interest.

The target genes can include both coding and non-coding regions. In particular embodiments, the trait is stress tolerance and the method is a method for the generation of stress-tolerant crop varieties

[00691] Use of AD-functionalized CRISPR to affect fruit-ripening

[00692] Ripening is a normal phase in the maturation process of fruits and vegetables. Only a few days after it starts it renders a fruit or vegetable inedible. This process brings significant losses to both farmers and consumers. In particular embodiments, the methods of the present invention are used to reduce ethylene production. This is ensured by ensuring one or more of the following: a. Suppression of ACC synthase gene expression. ACC (1-aminocyclopropane-1-carboxylic acid) synthase is the enzyme responsible for the conversion of S-adenosylmethionine (SAM) to ACC; the second to the last step in ethylene biosynthesis. Enzyme expression is hindered when an antisense ("mirror-image") or truncated copy of the synthase gene is inserted into the plant's genome; b. Insertion of the ACC deaminase gene. The gene coding for the enzyme is obtained from Pseudomonas chlororaphis, a common nonpathogenic soil bacterium. It converts ACC to a different compound thereby reducing the amount of ACC available for ethylene production; c. Insertion of the SAM hydrolase gene. This approach is similar to ACC deaminase wherein ethylene production is hindered when the amount of its precursor metabolite is reduced; in this case SAM is converted to homoserine. The gene coding for the enzyme is obtained from E. coli T3 bacteriophage and d. Suppression of ACC oxidase gene expression. ACC oxidase is the enzyme which catalyzes the oxidation of ACC to ethylene, the last step in the ethylene biosynthetic pathway. Using the methods described herein, down regulation of the ACC oxidase gene results in the suppression of ethylene production, thereby delaying fruit ripening. In particular embodiments, additionally or alternatively to the modifications described above, the methods described herein are used to modify ethylene receptors, so as to interfere with ethylene signals obtained by the fruit. In particular embodiments, expression of the ETR1 gene, encoding an ethylene binding protein is modified, more particularly suppressed. In particular embodiments, additionally or alternatively to the modifications described above, the methods described herein are used to modify expression of the gene encoding Polygalacturonase (PG), which is the enzyme responsible for the breakdown of pectin, the substance that maintains the integrity of plant cell walls. Pectin breakdown occurs at the start of the ripening process resulting in the softening of the fruit. Accordingly, in particular embodiments, the methods described herein are used to introduce

a mutation in the PG gene or to suppress activation of the PG gene in order to reduce the amount of PG enzyme produced thereby delaying pectin degradation.

[00693] Thus in particular embodiments, the methods comprise the use of the AD-functionalized CRISPR system to ensure one or more modifications of the genome of a plant cell such as described above, and regenerating a plant therefrom. In particular embodiments, the plant is a tomato plant.

[00694] Increasing storage life of plants

[00695] In particular embodiments, the methods of the present invention are used to modify genes involved in the production of compounds which affect storage life of the plant or plant part. More particularly, the modification is in a gene that prevents the accumulation of reducing sugars in potato tubers. Upon high-temperature processing, these reducing sugars react with free amino acids, resulting in brown, bitter-tasting products and elevated levels of acrylamide, which is a potential carcinogen. In particular embodiments, the methods provided herein are used to reduce or inhibit expression of the vacuolar invertase gene (VInv), which encodes a protein that breaks down sucrose to glucose and fructose (Clasen et al. DOI: 10.1111/pbi.12370).

[00696] The use of the AD-functionalized CRISPR system to ensure a value added trait

[00697] In particular embodiments the AD-functionalized CRISPR system is used to produce nutritionally improved agricultural crops. In particular embodiments, the methods provided herein are adapted to generate "functional foods", i.e. a modified food or food ingredient that may provide a health benefit beyond the traditional nutrients it contains and or "nutraceutical", i.e. substances that may be considered a food or part of a food and provides health benefits, including the prevention and treatment of disease. In particular embodiments, the nutraceutical is useful in the prevention and/or treatment of one or more of cancer, diabetes, cardiovascular disease, and hypertension.

[00698] Examples of nutritionally improved crops include (Newell-McGloughlin, Plant Physiology, July 2008, Vol. 147, pp. 939–953):

[00699] Modified protein quality, content and/or amino acid composition, such as have been described for Bahiagrass (Luciani et al. 2005, Florida Genetics Conference Poster), Canola (Roesler et al., 1997, Plant Physiol 113 75–81), Maize (Cromwell et al, 1967, 1969 J Anim Sci 26 1325–1331, O'Quin et al. 2000 J Anim Sci 78 2144–2149, Yang et al. 2002, Transgenic Res 11 11–20, Young et al. 2004, Plant J 38 910–922), Potato (Yu J and Ao, 1997 Acta Bot Sin 39 329–334; Chakraborty et al. 2000, Proc Natl Acad Sci USA 97 3724–

3729; Li et al. 2001) Chin Sci Bull 46 482–484, Rice (Katsube et al. 1999, Plant Physiol 120 1063–1074), Soybean (Dinkins et al. 2001, Rapp 2002, In Vitro Cell Dev Biol Plant 37 742–747), Sweet Potato (Egnin and Prakash 1997, In Vitro Cell Dev Biol 33 52A).

[00700] Essential amino acid content, such as has been described for Canola (Falco et al. 1995, Bio/Technology 13 577–582), Lupin (White et al. 2001, J Sci Food Agric 81 147–154), Maize (Lai and Messing, 2002, Agbios 2008 GM crop database (March 11, 2008)), Potato (Zeh et al. 2001, Plant Physiol 127 792–802), Sorghum (Zhao et al. 2003, Kluwer Academic Publishers, Dordrecht, The Netherlands, pp 413–416), Soybean (Falco et al. 1995 Bio/Technology 13 577–582; Galili et al. 2002 Crit Rev Plant Sci 21 167–204).

[00701] Oils and Fatty acids such as for Canola (Dehesh et al. (1996) Plant J 9 167–172 [PubMed] ; Del Vecchio (1996) INFORM International News on Fats, Oils and Related Materials 7 230–243; Roesler et al. (1997) Plant Physiol 113 75–81 [PMC free article] [PubMed]; Froman and Ursin (2002, 2003) Abstracts of Papers of the American Chemical Society 223 U35; James et al. (2003) Am J Clin Nutr 77 1140–1145 [PubMed]; Agbios (2008, above); coton (Chapman et al. (2001) . J Am Oil Chem Soc 78 941–947; Liu et al. (2002) J Am Coll Nutr 21 205S–211S [PubMed]; O'Neill (2007) Australian Life Scientist. http://www.biotechnews.com.au/index.php/id;866694817;fp;4;fpid;2 (June 17, 2008), Linseed (Abbadi et al., 2004, Plant Cell 16: 2734–2748), Maize (Young et al., 2004, Plant J 38 910–922), oil palm (Jalani et al. 1997, J Am Oil Chem Soc 74 1451–1455; Parveez, 2003, AgBiotechNet 113 1–8), Rice (Anai et al., 2003, Plant Cell Rep 21 988–992), Soybean (Reddy and Thomas, 1996, Nat Biotechnol 14 639–642; Kinney and Kwolton, 1998, Blackie Academic and Professional, London, pp 193–213), Sunflower (Arcadia, Biosciences 2008)

[00702] Carbohydrates, such as Fructans described for Chicory (Smeekens (1997) Trends Plant Sci 2 286–287, Sprenger et al. (1997) FEBS Lett 400 355–358, Sévenier et al. (1998) Nat Biotechnol 16 843–846), Maize (Caimi et al. (1996) Plant Physiol 110 355–363), Potato (Hellwege et al. ,1997 Plant J 12 1057–1065), Sugar Beet (Smeekens et al. 1997, above), Inulin, such as described for Potato (Hellewege et al. 2000, Proc Natl Acad Sci USA 97 8699–8704), Starch, such as described for Rice (Schwall et al. (2000) Nat Biotechnol 18 551–554, Chiang et al. (2005) Mol Breed 15 125–143),

[00703] Vitamins and carotenoids, such as described for Canola (Shintani and DellaPenna (1998) Science 282 2098–2100), Maize (Rocheford et al. (2002) . J Am Coll Nutr 21 191S–198S, Cahoon et al. (2003) Nat Biotechnol 21 1082–1087, Chen et al. (2003) Proc Natl Acad Sci USA 100 3525–3530), Mustardseed (Shewmaker et al. (1999) Plant J 20

401–412, Potato (Ducreux et al., 2005, J Exp Bot 56 81–89), Rice (Ye et al. (2000) Science 287 303–305, Strawberry (Agius et al. (2003), Nat Biotechnol 21 177–181), Tomato (Rosati et al. (2000) Plant J 24 413–419, Fraser et al. (2001) J Sci Food Agric 81 822–827, Mehta et al. (2002) Nat Biotechnol 20 613–618, Díaz de la Garza et al. (2004) Proc Natl Acad Sci USA 101 13720–13725, Enfissi et al. (2005) Plant Biotechnol J 3 17–27, DellaPenna (2007) Proc Natl Acad Sci USA 104 3675–3676.

[00704] Functional secondary metabolites, such as described for Apple (stilbenes, Szankowski et al. (2003) Plant Cell Rep 22: 141–149), Alfalfa (resveratrol, Hipskind and Paiva (2000) Mol Plant Microbe Interact 13 551–562), Kiwi (resveratrol, Kobayashi et al. (2000) Plant Cell Rep 19 904–910), Maize and Soybean (flavonoids, Yu et al. (2000) Plant Physiol 124 781–794), Potato (anthocyanin and alkaloid glycoside, Lukaszewicz et al. (2004) J Agric Food Chem 52 1526–1533), Rice (flavonoids & resveratrol, Stark-Lorenzen et al. (1997) Plant Cell Rep 16 668–673, Shin et al. (2006) Plant Biotechnol J 4 303–315), Tomato (+resveratrol, chlorogenic acid, flavonoids, stilbene; Rosati et al. (2000) above, Muir et al. (2001) Nature 19 470–474, Niggeweg et al. (2004) Nat Biotechnol 22 746–754, Giovinazzo et al. (2005) Plant Biotechnol J 3 57–69), wheat (caffeic and ferulic acids, resveratrol; United Press International (2002)); and

[00705] Mineral availabilities such as described for Alfalfa (phytase, Austin-Phillips et al. (1999) http://www.molecularfarming.com/nonmedical.html), Lettuse (iron, Goto et al. (2000) Theor Appl Genet 100 658–664), Rice (iron, Lucca et al. (2002) J Am Coll Nutr 21 184S–190S), Maize, Soybean and wheate (phytase, Drakakaki et al. (2005) Plant Mol Biol 59 869–880, Denbow et al. (1998) Poult Sci 77 878–881, Brinch-Pedersen et al. (2000) Mol Breed 6 195–206).

[00706] In particular embodiments, the value-added trait is related to the envisaged health benefits of the compounds present in the plant. For instance, in particular embodiments, the value-added crop is obtained by applying the methods of the invention to ensure the modification of or induce/increase the synthesis of one or more of the following compounds:

[00707] Carotenoids, such as α-Carotene present in carrots which Neutralizes free radicals that may cause damage to cells or β-Carotene present in various fruits and vegetables which neutralizes free radicals

[00708] Lutein present in green vegetables which contributes to maintenance of healthy vision

[00709] Lycopene present in tomato and tomato products, which is believed to reduce the risk of prostate cancer

[00710] Zeaxanthin, present in citrus and maize, which contributes to mainteance of healthy vision

[00711] Dietary fiber such as insoluble fiber present in wheat bran which may reduce the risk of breast and/or colon cancer and β-Glucan present in oat, soluble fiber present in Psylium and whole cereal grains which may reduce the risk of cardiovascular disease (CVD)

[00712] Fatty acids, such as ω-3 fatty acids which may reduce the risk of CVD and improve mental and visual functions, Conjugated linoleic acid, which may improve body composition, may decrease risk of certain cancers and GLA which may reduce inflammation risk of cancer and CVD, may improve body composition

[00713] Flavonoids such as Hydroxycinnamates, present in wheat which have Antioxidant-like activities, may reduce risk of degenerative diseases, flavonols, catechins and tannins present in fruits and vegetables which neutralize free radicals and may reduce risk of cancer

[00714] Glucosinolates, indoles, isothiocyanates, such as Sulforaphane, present in Cruciferous vegetables (broccoli, kale), horseradish, which neutralize free radicals, may reduce risk of cancer

[00715] Phenolics, such as stilbenes present in grape which May reduce risk of degenerative diseases, heart disease, and cancer, may have longevity effect and caffeic acid and ferulic acid present in vegetables and citrus which have Antioxidant-like activities, may reduce risk of degenerative diseases, heart disease, and eye disease, and epicatechin present in cacao which has Antioxidant-like activities, may reduce risk of degenerative diseases and heart disease

[00716] Plant stanols/sterols present in maize, soy, wheat and wooden oils which May reduce risk of coronary heart disease by lowering blood cholesterol levels

[00717] Fructans, inulins, fructo-oligosaccharides present in Jerusalem artichoke, shallot, onion powder which may improve gastrointestinal health

[00718] Saponins present in soybean, which may lower LDL cholesterol

[00719] Soybean protein present in soybean which may reduce risk of heart disease

[00720] Phytoestrogens such as isoflavones present in soybean which May reduce menopause symptoms, such as hot flashes, may reduce osteoporosis and CVD and lignans

present in flax, rye and vegetables, which May protect against heart disease and some cancers, may lower LDL cholesterol, total cholesterol.

[00721] Sulfides and thiols such as diallyl sulphide present in onion, garlic, olive, leek and scallon and Allyl methyl trisulfide, dithiolthiones present in cruciferous vegetables which may lower LDL cholesterol, helps to maintain healthy immune system

[00722] Tannins, such as proanthocyanidins, present in cranberry, cocoa, which may improve urinary tract health, may reduce risk of CVD and high blood pressure.

[00723] In addition, the methods of the present invention also envisage modifying protein/starch functionality, shelf life, taste/aesthetics, fiber quality, and allergen, antinutrient, and toxin reduction traits.

[00724] Accordingly, the invention encompasses methods for producing plants with nutritional added value, said methods comprising introducing into a plant cell a gene encoding an enzyme involved in the production of a component of added nutritional value using the AD-functionalized CRISPR system as described herein and regenerating a plant from said plant cell, said plant characterized in an increase expression of said component of added nutritional value. In particular embodiments, the AD-functionalized CRISPR system is used to modify the endogenous synthesis of these compounds indirectly, e.g. by modifying one or more transcription factors that controls the metabolism of this compound. Methods for introducing a gene of interest into a plant cell and/or modifying an endogenous gene using the AD-functionalized CRISPR system are described herein above.

[00725] Some specific examples of modifications in plants that have been modified to confer value-added traits are: plants with modified fatty acid metabolism, for example, by transforming a plant with an antisense gene of stearyl-ACP desaturase to increase stearic acid content of the plant. See Knultzon et al., Proc. Natl. Acad. Sci. U.S.A. 89:2624 (1992). Another example involves decreasing phytate content, for example by cloning and then reintroducing DNA associated with the single allele which may be responsible for maize mutants characterized by low levels of phytic acid. See Raboy et al, Maydica 35:383 (1990).

[00726] Similarly, expression of the maize (Zea mays) Tfs C1 and R, which regulate the production of flavonoids in maize aleurone layers under the control of a strong promoter, resulted in a high accumulation rate of anthocyanins in Arabidopsis (Arabidopsis thaliana), presumably by activating the entire pathway (Bruce et al., 2000, Plant Cell 12:65–80). DellaPenna (Welsch et al., 2007 Annu Rev Plant Biol 57: 711–738) found that Tf RAP2.2 and its interacting partner SINAT2 increased carotenogenesis in Arabidopsis leaves.

Expressing the Tf Dof1 induced the up-regulation of genes encoding enzymes for carbon skeleton production, a marked increase of amino acid content, and a reduction of the Glc level in transgenic Arabidopsis (Yanagisawa, 2004 Plant Cell Physiol 45: 386–391), and the DOF Tf AtDof1.1 (OBP2) up-regulated all steps in the glucosinolate biosynthetic pathway in Arabidopsis (Skirycz et al., 2006 Plant J 47: 10–24).

[00727] Reducing allergen in plants

[00728] In particular embodiments the methods provided herein are used to generate plants with a reduced level of allergens, making them safer for the consumer. In particular embodiments, the methods comprise modifying expression of one or more genes responsible for the production of plant allergens. For instance, in particular embodiments, the methods comprise down-regulating expression of a Lol p5 gene in a plant cell, such as a ryegrass plant cell and regenerating a plant therefrom so as to reduce allergenicity of the pollen of said plant (Bhalla et al. 1999, Proc. Natl. Acad. Sci. USA Vol. 96: 11676–11680).

[00729] Peanut allergies and allergies to legumes generally are a real and serious health concern. The AD-functionalized CRISPR system of the present invention can be used to identify and then mutate genes encoding allergenic proteins of such legumes. Without limitation as to such genes and proteins, Nicolaou et al. identifies allergenic proteins in peanuts, soybeans, lentils, peas, lupin, green beans, and mung beans. See, Nicolaou et al., Current Opinion in Allergy and Clinical Immunology 2011;11(3):222).

[00730] Screening methods for endogenous genes of interest

[00731] The methods provided herein further allow the identification of genes of value encoding enzymes involved in the production of a component of added nutritional value or generally genes affecting agronomic traits of interest, across species, phyla, and plant kingdom. By selectively targeting e.g. genes encoding enzymes of metabolic pathways in plants using the AD-functionalized CRISPR system as described herein, the genes responsible for certain nutritional aspects of a plant can be identified. Similarly, by selectively targeting genes which may affect a desirable agronomic trait, the relevant genes can be identified. Accordingly, the present invention encompasses screening methods for genes encoding enzymes involved in the production of compounds with a particular nutritional value and/or agronomic traits.

[00732] Further applications of the AD-functionalized CRISPR system in plants and yeasts

[00733] Use of AD-functionalized CRISPR system in biofuel production

[00734] The term "biofuel" as used herein is an alternative fuel made from plant and plant-derived resources. Renewable biofuels can be extracted from organic matter whose energy has been obtained through a process of carbon fixation or are made through the use or conversion of biomass. This biomass can be used directly for biofuels or can be converted to convenient energy containing substances by thermal conversion, chemical conversion, and biochemical conversion. This biomass conversion can result in fuel in solid, liquid, or gas form. There are two types of biofuels: bioethanol and biodiesel. Bioethanol is mainly produced by the sugar fermentation process of cellulose (starch), which is mostly derived from maize and sugar cane. Biodiesel on the other hand is mainly produced from oil crops such as rapeseed, palm, and soybean. Biofuels are used mainly for transportation.

[00735] **Enhancing plant properties for biofuel production**

[00736] In particular embodiments, the methods using the AD-functionalized CRISPR system as described herein are used to alter the properties of the cell wall in order to facilitate access by key hydrolysing agents for a more efficient release of sugars for fermentation. In particular embodiments, the biosynthesis of cellulose and/or lignin are modified. Cellulose is the major component of the cell wall. The biosynthesis of cellulose and lignin are co-regulated. By reducing the proportion of lignin in a plant the proportion of cellulose can be increased. In particular embodiments, the methods described herein are used to downregulate lignin biosynthesis in the plant so as to increase fermentable carbohydrates. More particularly, the methods described herein are used to downregulate at least a first lignin biosynthesis gene selected from the group consisting of 4-coumarate 3-hydroxylase (C3H), phenylalanine ammonia-lyase (PAL), cinnamate 4-hydroxylase (C4H), hydroxycinnamoyl transferase (HCT), caffeic acid O-methyltransferase (COMT), caffeoyl CoA 3-O-methyltransferase (CCoAOMT), ferulate 5- hydroxylase (F5H), cinnamyl alcohol dehydrogenase (CAD), cinnamoyl CoA-reductase (CCR), 4- coumarate-CoA ligase (4CL), monolignol-lignin-specific glycosyltransferase, and aldehyde dehydrogenase (ALDH) as disclosed in WO 2008064289 A2.

[00737] In particular embodiments, the methods described herein are used to produce plant mass that produces lower levels of acetic acid during fermentation (see also WO 2010096488). More particularly, the methods disclosed herein are used to generate mutations in homologs to CaslL to reduce polysaccharide acetylation.

[00738] **Modifying yeast for Biofuel production**

[00739] In particular embodiments, the AD-functionalized CRISPR system provided herein is used for bioethanol production by recombinant micro-organisms. For instance, the

AD-functionalized CRISPR system can be used to engineer micro-organisms, such as yeast, to generate biofuel or biopolymers from fermentable sugars and optionally to be able to degrade plant-derived lignocellulose derived from agricultural waste as a source of fermentable sugars. In some embodiments, the AD-functionalized CRISPR system is used to modify endogenous metabolic pathways which compete with the biofuel production pathway.

[00740] Accordingly, in more particular embodiments, the methods described herein are used to modify a micro-organism as follows: to modify at least one nucleic acid encoding for an enzyme in a metabolic pathway in said host cell, wherein said pathway produces a metabolite other than acetaldehyde from pyruvate or ethanol from acetaldehyde, and wherein said modification results in a reduced production of said metabolite, or to introduce at least one nucleic acid encoding for an inhibitor of said enzyme.

[00741] **Modifying Algae and plants for production of vegetable oils or biofuels**

[00742] Transgenic algae or other plants such as rape may be particularly useful in the production of vegetable oils or biofuels such as alcohols (especially methanol and ethanol), for instance. These may be engineered to express or overexpress high levels of oil or alcohols for use in the oil or biofuel industries.

[00743] According to particular embodiments of the invention, the AD-functionalized CRISPR system is used to generate lipid-rich diatoms which are useful in biofuel production.

[00744] In particular embodiments it is envisaged to specifically modify genes that are involved in the modification of the quantity of lipids and/or the quality of the lipids produced by the algal cell. Examples of genes encoding enzymes involved in the pathways of fatty acid synthesis can encode proteins having for instance acetyl-CoA carboxylase, fatty acid synthase, 3-ketoacyl_acyl- carrier protein synthase III, glycerol-3-phospate deshydrogenase (G3PDH), Enoyl-acyl carrier protein reductase (Enoyl-ACP-reductase), glycerol-3-phosphate acyltransferase, lysophosphatidic acyl transferase or diacylglycerol acyltransferase, phospholipid:diacylglycerol acyltransferase, phoshatidate phosphatase, fatty acid thioesterase such as palmitoyi protein thioesterase, or malic enzyme activities. In further embodiments it is envisaged to generate diatoms that have increased lipid accumulation. This can be achieved by targeting genes that decrease lipid catabolisation. Of particular interest for use in the methods of the present invention are genes involved in the activation of both triacylglycerol and free fatty acids, as well as genes directly involved in β-oxidation of fatty acids, such as acyl-CoA synthetase, 3-ketoacyl-CoA thiolase, acyl-CoA

oxidase activity and phosphoglucomutase. The AD-functionalized CRISPR system and methods described herein can be used to specifically activate such genes in diatoms as to increase their lipid content.

[00745] Organisms such as microalgae are widely used for synthetic biology. Stovicek et al. (Metab. Eng. Comm., 2015; 2:13 describes genome editing of industrial yeast, for example, *Saccharomyces cerevisae*, to efficiently produce robust strains for industrial production. Stovicek used a CRISPR-Cas9 system codon-optimized for yeast to simultaneously disrupt both alleles of an endogenous gene and knock in a heterologous gene. Cas9 and guide RNA were expressed from genomic or episomal 2μ-based vector locations. The authors also showed that gene disruption efficiency could be improved by optimization of the levels of Cas9 and guide RNA expression. Hlavová et al. (Biotechnol. Adv. 2015) discusses development of species or strains of microalgae using techniques such as CRISPR to target nuclear and chloroplast genes for insertional mutagenesis and screening.

[00746] US 8945839 describes a method for engineering Micro-Algae (Chlamydomonas reinhardtii cells) species) using Cas9 . Using similar tools, the methods of the AD-functionalized CRISPR system described herein can be applied on Chlamydomonas species and other algae. In particular embodiments, a CRISPR-Cas protein (e.g., Cpf1), adenosine deaminase (which may be fused to the CRISPR-Cas protein or an aptamer-binding adaptor protein), and guide RNA are introduced in algae expressed using a vector that expresses the CRISPR-Cas protein and optionally the adenosine deaminase under the control of a constitutive promoter such as Hsp70A-Rbc S2 or Beta2 -tubulin. Guide RNA will be delivered using a vector containing T7 promoter. Alternatively, mRNA and in vitro transcribed guide RNA can be delivered to algal cells. Electroporation protocol follows standard recommended protocol from the GeneArt Chlamydomonas Engineering kit.

[00747] The use of AD-functionalized CRISPR system in the generation of micro-organisms capable of fatty acid production

[00748] In particular embodiments, the methods of the invention are used for the generation of genetically engineered micro-organisms capable of the production of fatty esters, such as fatty acid methyl esters ("FAME") and fatty acid ethyl esters ("FAEE"),

[00749] Typically, host cells can be engineered to produce fatty esters from a carbon source, such as an alcohol, present in the medium, by expression or overexpression of a gene encoding a thioesterase, a gene encoding an acyl-CoA synthase, and a gene encoding an ester synthase. Accordingly, the methods provided herein are used to modify a micro-

organisms so as to overexpress or introduce a thioesterase gene, a gene encloding an acyl-CoA synthase, and a gene encoding an ester synthase. In particular embodiments, the thioesterase gene is selected from tesA, 'tesA, tesB,fatB, fatB2,fatB3,fatAl, or fatA. In particular embodiments, the gene encoding an acyl-CoA synthase is selected from fadDJadK, BH3103, pfl-4354, EAV15023, fadDl, fadD2, RPC_4074,fadDD35, fadDD22, faa39, or an identified gene encoding an enzyme having the same properties. In particular embodiments, the gene encoding an ester synthase is a gene encoding a synthase/acyl-CoA:diacylglycerl acyltransferase from Simmondsia chinensis, Acinetobacter sp. ADP , Alcanivorax borkumensis, Pseudomonas aeruginosa, Fundibacter jadensis, Arabidopsis thaliana, or Alkaligenes eutrophus, or a variant thereof. Additionally or alternatively, the methods provided herein are used to decrease expression in said micro-organism of of at least one of a gene encoding an acyl-CoA dehydrogenase, a gene encoding an outer membrane protein receptor, and a gene encoding a transcriptional regulator of fatty acid biosynthesis. In particular embodiments one or more of these genes is inactivated, such as by introduction of a mutation. In particular embodiments, the gene encoding an acyl-CoA dehydrogenase is fadE. In particular embodiments, the gene encoding a transcriptional regulator of fatty acid biosynthesis encodes a DNA transcription repressor, for example, fabR.

[00750] Additionally or alternatively, said micro-organism is modified to reduce expression of at least one of a gene encoding a pyruvate formate lyase, a gene encoding a lactate dehydrogenase, or both. In particular embodiments, the gene encoding a pyruvate formate lyase is pflB. In particular embodiments, the gene encoding a lactate dehydrogenase is IdhA. In particular embodiments one or more of these genes is inactivated, such as by introduction of a mutation therein.

[00751] In particular embodiments, the micro-organism is selected from the genus *Escherichia, Bacillus, Lactobacillus, Rhodococcus, Synechococcus, Synechoystis, Pseudomonas, Aspergillus, Trichoderma, Neurospora, Fusarium, Humicola, Rhizomucor, Kluyveromyces, Pichia, Mucor, Myceliophtora, Penicillium, Phanerochaete, Pleurotus, Trametes, Chrysosporium, Saccharomyces, Stenotrophamonas, Schizosaccharomyces, Yarrowia, or Streptomyces.*

[00752] The use of AD-functionalized CRISPR system in the generation of micro-organisms capable of organic acid production

[00753] The methods provided herein are further used to engineer micro-organisms capable of organic acid production, more particularly from pentose or hexose sugars. In

particular embodiments, the methods comprise introducing into a micro-organism an exogenous LDH gene. In particular embodiments, the organic acid production in said micro-organisms is additionally or alternatively increased by inactivating endogenous genes encoding proteins involved in an endogenous metabolic pathway which produces a metabolite other than the organic acid of interest and/or wherein the endogenous metabolic pathway consumes the organic acid. In particular embodiments, the modification ensures that the production of the metabolite other than the organic acid of interest is reduced. According to particular embodiments, the methods are used to introduce at least one engineered gene deletion and/or inactivation of an endogenous pathway in which the organic acid is consumed or a gene encoding a product involved in an endogenous pathway which produces a metabolite other than the organic acid of interest. In particular embodiments, the at least one engineered gene deletion or inactivation is in one or more gene encoding an enzyme selected from the group consisting of pyruvate decarboxylase (pdc), fumarate reductase, alcohol dehydrogenase (adh), acetaldehyde dehydrogenase, phosphoenolpyruvate carboxylase (ppc), D-lactate dehydrogenase (d-ldh), L-lactate dehydrogenase (l-ldh), lactate 2-monooxygenase.
In further embodiments the at least one engineered gene deletion and/or inactivation is in an endogenous gene encoding pyruvate decarboxylase (pdc).

[00754] In further embodiments, the micro-organism is engineered to produce lactic acid and the at least one engineered gene deletion and/or inactivation is in an endogenous gene encoding lactate dehydrogenase. Additionally or alternatively, the micro-organism comprises at least one engineered gene deletion or inactivation of an endogenous gene encoding a cytochrome-dependent lactate dehydrogenase, such as a cytochrome B2-dependent L-lactate dehydrogenase.

[00755] The use of AD-functionalized CRISPR system in the generation of improved xylose or cellobiose utilizing yeasts strains

[00756] In particular embodiments, the AD-functionalized CRISPR system may be applied to select for improved xylose or cellobiose utilizing yeast strains. Error-prone PCR can be used to amplify one (or more) genes involved in the xylose utilization or cellobiose utilization pathways. Examples of genes involved in xylose utilization pathways and cellobiose utilization pathways may include, without limitation, those described in Ha, S.J., et al. (2011) Proc. Natl. Acad. Sci. USA 108(2):504-9 and Galazka, J.M., et al. (2010) Science 330(6000):84-6. Resulting libraries of double-stranded DNA molecules, each comprising a random mutation in such a selected gene could be co-transformed with the

components of the AD-functionalized CRISPR system into a yeast strain (for instance S288C) and strains can be selected with enhanced xylose or cellobiose utilization capacity, as described in WO2015138855.

[00757] The use of AD-functionalized CRISPR system in the generation of improved yeasts strains for use in isoprenoid biosynthesis

[00758] Tadas Jakočiūnas et al. described the successful application of a multiplex CRISPR-Cas9 system for genome engineering of up to 5 different genomic loci in one transformation step in baker's yeast Saccharomyces cerevisiae (Metabolic Engineering Volume 28, March 2015, Pages 213–222) resulting in strains with high mevalonate production, a key intermediate for the industrially important isoprenoid biosynthesis pathway. In particular embodiments, the AD-functionalized CRISPR system may be applied in a multiplex genome engineering method as described herein for identifying additional high producing yeast strains for use in isoprenoid synthesis.

[00759] Improved plants and yeast cells

[00760] The present invention also provides plants and yeast cells obtainable and obtained by the methods provided herein. The improved plants obtained by the methods described herein may be useful in food or feed production through expression of genes which, for instance ensure tolerance to plant pests, herbicides, drought, low or high temperatures, excessive water, etc.

[00761] The improved plants obtained by the methods described herein, especially crops and algae may be useful in food or feed production through expression of, for instance, higher protein, carbohydrate, nutrient or vitamin levels than would normally be seen in the wildtype. In this regard, improved plants, especially pulses and tubers are preferred.

[00762] Improved algae or other plants such as rape may be particularly useful in the production of vegetable oils or biofuels such as alcohols (especially methanol and ethanol), for instance. These may be engineered to express or overexpress high levels of oil or alcohols for use in the oil or biofuel industries.

[00763] The invention also provides for improved parts of a plant. Plant parts include, but are not limited to, leaves, stems, roots, tubers, seeds, endosperm, ovule, and pollen. Plant parts as envisaged herein may be viable, nonviable, regeneratable, and/or non-regeneratable.

[00764] It is also encompassed herein to provide plant cells and plants generated according to the methods of the invention. Gametes, seeds, embryos, either zygotic or somatic, progeny or hybrids of plants comprising the genetic modification, which are

produced by traditional breeding methods, are also included within the scope of the present invention. Such plants may contain a heterologous or foreign DNA sequence inserted at or instead of a target sequence. Alternatively, such plants may contain only an alteration (mutation, deletion, insertion, substitution) in one or more nucleotides. As such, such plants will only be different from their progenitor plants by the presence of the particular modification.

[00765] Thus, the invention provides a plant, animal or cell, produced by the present methods, or a progeny thereof. The progeny may be a clone of the produced plant or animal, or may result from sexual reproduction by crossing with other individuals of the same species to introgress further desirable traits into their offspring. The cell may be *in vivo* or *ex vivo* in the cases of multicellular organisms, particularly animals or plants.

[00766] The methods for genome editing using the AD-functionalized CRISPR system as described herein can be used to confer desired traits on essentially any plant, algae, fungus, yeast, etc. A wide variety of plants, algae, fungus, yeast, etc and plant algae, fungus, yeast cell or tissue systems may be engineered for the desired physiological and agronomic characteristics described herein using the nucleic acid constructs of the present disclosure and the various transformation methods mentioned above.

[00767] In particular embodiments, the methods described herein are used to modify endogenous genes or to modify their expression without the permanent introduction into the genome of the plant, algae, fungus, yeast, etc of any foreign gene, including those encoding CRISPR components, so as to avoid the presence of foreign DNA in the genome of the plant. This can be of interest as the regulatory requirements for non-transgenic plants are less rigorous.

[00768] The methods described herein generally result in the generation of "improved plants, algae, fungi, yeast, etc" in that they have one or more desirable traits compared to the wildtype plant. In particular embodiments, non-transgenic genetically modified plants, algae, fungi, yeast, etc., parts or cells are obtained, in that no exogenous DNA sequence is incorporated into the genome of any of the cells of the plant. In such embodiments, the improved plants, algae, fungi, yeast, etc. are non-transgenic. Where only the modification of an endogenous gene is ensured and no foreign genes are introduced or maintained in the plant, algae, fungi, yeast, etc. genome, the resulting genetically modified crops contain no foreign genes and can thus basically be considered non-transgenic. The different applications of the AD-functionalized CRISPR system for plant, algae, fungi, yeast, etc. genome editing include, but are not limited to: editing of endogenous genes to confer an

agricultural trait of interest. Examplary genes conferring agronomic traits include, but are not limited to genes that confer resistance to pests or diseases; genes involved in plant diseases, such as those listed in WO 2013046247; genes that confer resistance to herbicides, fungicides, or the like; genes involved in (abiotic) stress tolerance. Other aspects of the use of the CRISPR-Cas system include, but are not limited to: create (male) sterile plants; increasing the fertility stage in plants/algae etc; generate genetic variation in a crop of interest; affect fruit-ripening; increasing storage life of plants/algae etc; reducing allergen in plants/algae etc; ensure a value added trait (e.g. nutritional improvement); Sscreening methods for endogenous genes of interest; biofuel, fatty acid, organic acid, etc production.

[00769] AD-Functionalized CRISPR System Can Be Used In Non-Human Organisms

[00770] In an aspect, the invention provides a non-human eukaryotic organism; preferably a multicellular eukaryotic organism, comprising a eukaryotic host cell according to any of the described embodiments. In other aspects, the invention provides a eukaryotic organism; preferably a multicellular eukaryotic organism, comprising a eukaryotic host cell according to any of the described embodiments. The organism in some embodiments of these aspects may be an animal; for example a mammal. Also, the organism may be an arthropod such as an insect. The present invention may also be extended to other agricultural applications such as, for example, farm and production animals. For example, pigs have many features that make them attractive as biomedical models, especially in regenerative medicine. In particular, pigs with severe combined immunodeficiency (SCID) may provide useful models for regenerative medicine, xenotransplantation (discussed also elsewhere herein), and tumor development and will aid in developing therapies for human SCID patients. Lee et al., (Proc Natl Acad Sci U S A. 2014 May 20;111(20):7260-5) utilized a reporter-guided transcription activator-like effector nuclease (TALEN) system to generated targeted modifications of recombination activating gene (RAG) 2 in somatic cells at high efficiency, including some that affected both alleles. The AD-functionalized CRISPR system may be applied to a similar system.

[00771] The methods of Lee et al., (Proc Natl Acad Sci U S A. 2014 May 20;111(20):7260-5) may be applied to the present invention analogously as follows. Mutated pigs are produced by targeted modification of RAG2 in fetal fibroblast cells followed by SCNT and embryo transfer. Constructs coding for CRISPR Cas and a reporter are electroporated into fetal-derived fibroblast cells. After 48 h, transfected cells expressing the green fluorescent protein are sorted into individual wells of a 96-well plate at an

estimated dilution of a single cell per well. Targeted modification of RAG2 are screened by amplifying a genomic DNA fragment flanking any CRISPR Cas cutting sites followed by sequencing the PCR products. After screening and ensuring lack of off-site mutations, cells carrying targeted modification of RAG2 are used for SCNT. The polar body, along with a portion of the adjacent cytoplasm of oocyte, presumably containing the metaphase II plate, are removed, and a donor cell are placed in the perivitelline. The reconstructed embryos are then electrically porated to fuse the donor cell with the oocyte and then chemically activated. The activated embryos are incubated in Porcine Zygote Medium 3 (PZM3) with 0.5 μM Scriptaid (S7817; Sigma-Aldrich) for 14–16 h. Embryos are then washed to remove the Scriptaid and cultured in PZM3 until they were transferred into the oviducts of surrogate pigs.

[00772] The present invention is used to create a platform to model a disease or disorder of an animal, in some embodiments a mammal, in some embodiments a human. In certain embodiments, such models and platforms are rodent based, in non-limiting examples rat or mouse. Such models and platforms can take advantage of distinctions among and comparisons between inbred rodent strains. In certain embodiments, such models and platforms primate, horse, cattle, sheep, goat, swine, dog, cat or bird-based, for example to directly model diseases and disorders of such animals or to create modified and/or improved lines of such animals. Advantageously, in certain embodiments, an animal based platform or model is created to mimic a human disease or disorder. For example, the similarities of swine to humans make swine an ideal platform for modeling human diseases. Compared to rodent models, development of swine models has been costly and time intensive. On the other hand, swine and other animals are much more similar to humans genetically, anatomically, physiologically and pathophysiologically. The present invention provides a high efficiency platform for targeted gene and genome editing, gene and genome modification and gene and genome regulation to be used in such animal platforms and models. Though ethical standards block development of human models and in many cases models based on non-human primates, the present invention is used with in vitro systems, including but not limited to cell culture systems, three dimensional models and systems, and organoids to mimic, model, and investigate genetics, anatomy, physiology and pathophysiology of structures, organs, and systems of humans. The platforms and models provide manipulation of single or multiple targets.

[00773] In certain embodiments, the present invention is applicable to disease models like that of Schomberg et al. (FASEB Journal, April 2016; 30(1):Suppl 571.1). To model

the inherited disease neurofibromatosis type 1 (NF-1) Schomberg used CRISPR-Cas9 to introduce mutations in the swine neurofibromin 1 gene by cytosolic microinjection of CRISPR/Cas9 components into swine embryos. CRISPR guide RNAs (gRNA) were created for regions targeting sites both upstream and downstream of an exon within the gene for targeted cleavage by Cas9 and repair was mediated by a specific single-stranded oligodeoxynucleotide (ssODN) template to introduce a 2500 bp deletion. The CRISPR-Cas system was also used to engineer swine with specific NF-1 mutations or clusters of mutations, and futher can be used to engineer mutations that are specific to or representative of a given human individual. The invention is similarly used to develop animal models, including but not limited to swine models, of human multigenic diseases. According to the invention, multiple genetic loci in one gene or in multiple genes are simultaneously targeted using multiplexed guides and optionally one or multiple templates.

[00774] The present invention is also applicable to modifying SNPs of other animals, such as cows. Tan et al. (Proc Natl Acad Sci U S A. 2013 Oct 8; 110(41): 16526–16531) expanded the livestock gene editing toolbox to include transcription activator-like (TAL) effector nuclease (TALEN)- and clustered regularly interspaced short palindromic repeats (CRISPR)/Cas9- stimulated homology-directed repair (HDR) using plasmid, rAAV, and oligonucleotide templates. Gene specific guide RNA sequences were cloned into the Church lab guide RNA vector (Addgene ID: 41824) according to their methods (Mali P, et al. (2013) RNA-Guided Human Genome Engineering via Cas9. Science 339(6121):823-826). The Cas9 nuclease was provided either by co-transfection of the hCas9 plasmid (Addgene ID: 41815) or mRNA synthesized from RCIScript-hCas9. This RCIScript-hCas9 was constructed by sub-cloning the XbaI-AgeI fragment from the hCas9 plasmid (encompassing the hCas9 cDNA) into the RCIScript plasmid.

[00775] Heo et al. (Stem Cells Dev. 2015 Feb 1;24(3):393-402. doi: 10.1089/scd.2014.0278. Epub 2014 Nov 3) reported highly efficient gene targeting in the bovine genome using bovine pluripotent cells and clustered regularly interspaced short palindromic repeat (CRISPR)/Cas9 nuclease. First, Heo et al. generate induced pluripotent stem cells (iPSCs) from bovine somatic fibroblasts by the ectopic expression of yamanaka factors and GSK3β and MEK inhibitor (2i) treatment. Heo et al. observed that these bovine iPSCs are highly similar to naïve pluripotent stem cells with regard to gene expression and developmental potential in teratomas. Moreover, CRISPR-Cas9 nuclease, which was specific for the bovine NANOG locus, showed highly efficient editing of the bovine genome in bovine iPSCs and embryos.

[00776] Igenity® provides a profile analysis of animals, such as cows, to perform and transmit traits of economic traits of economic importance, such as carcass composition, carcass quality, maternal and reproductive traits and average daily gain. The analysis of a comprehensive Igenity® profile begins with the discovery of DNA markers (most often single nucleotide polymorphisms or SNPs). All the markers behind the Igenity® profile were discovered by independent scientists at research institutions, including universities, research organizations, and government entities such as USDA. Markers are then analyzed at Igenity® in validation populations. Igenity® uses multiple resource populations that represent various production environments and biological types, often working with industry partners from the seedstock, cow-calf, feedlot and/or packing segments of the beef industry to collect phenotypes that are not commonly available. Cattle genome databases are widely available, see, e.g., the NAGRP Cattle Genome Coordination Program (http://www.animalgenome.org/cattle/maps/db.html). Thus, the present invention maybe applied to target bovine SNPs. One of skill in the art may utilize the above protocols for targeting SNPs and apply them to bovine SNPs as described, for example, by Tan et al. or Heo et al.

[00777] Qingjian Zou et al. (Journal of Molecular Cell Biology Advance Access published October 12, 2015) demonstrated increased muscle mass in dogs by targeting targeting the first exon of the dog Myostatin (MSTN) gene (a negative regulator of skeletal muscle mass). First, the efficiency of the sgRNA was validated, using cotransfection of the the sgRNA targeting MSTN with a Cas9 vector into canine embryonic fibroblasts (CEFs). Thereafter, MSTN KO dogs were generated by micro-injecting embryos with normal morphology with a mixture of Cas9 mRNA and MSTN sgRNA and auto-transplantation of the zygotes into the oviduct of the same female dog. The knock-out puppies displayed an obvious muscular phenotype on thighs compared with its wild-type littermate sister. This can also be performed using the AD-functionalized CRISPR systems provided herein.

[00778] **Livestock - Pigs**

[00779] Viral targets in livestock may include, in some embodiments, porcine CD163, for example on porcine macrophages. CD163 is associated with infection (thought to be through viral cell entry) by PRRSv (Porcine Reproductive and Respiratory Syndrome virus, an arterivirus). Infection by PRRSv, especially of porcine alveolar macrophages (found in the lung), results in a previously incurable porcine syndrome ("Mystery swine disease" or "blue ear disease") that causes suffering, including reproductive failure, weight loss and high mortality rates in domestic pigs. Opportunistic infections, such as enzootic

pneumonia, meningitis and ear oedema, are often seen due to immune deficiency through loss of macrophage activity. It also has significant economic and environmental repercussions due to increased antibiotic use and financial loss (an estimated $660m per year).

[00780] As reported by Kristin M Whitworth and Dr Randall Prather et al. (Nature Biotech 3434 published online 07 December 2015) at the University of Missouri and in collaboration with Genus Plc, CD163 was targeted using CRISPR-Cas9 and the offspring of edited pigs were resistant when exposed to PRRSv. One founder male and one founder female, both of whom had mutations in exon 7 of CD163, were bred to produce offspring. The founder male possessed an 11-bp deletion in exon 7 on one allele, which results in a frameshift mutation and missense translation at amino acid 45 in domain 5 and a subsequent premature stop codon at amino acid 64. The other allele had a 2-bp addition in exon 7 and a 377-bp deletion in the preceding intron, which were predicted to result in the expression of the first 49 amino acids of domain 5, followed by a premature stop code at amino acid 85. The sow had a 7 bp addition in one allele that when translated was predicted to express the first 48 amino acids of domain 5, followed by a premature stop codon at amino acid 70. The sow's other allele was unamplifiable. Selected offspring were predicted to be a null animal (CD163–/–), i.e. a CD163 knock out.

[00781] Accordingly, in some embodiments, porcine alveolar macrophages may be targeted by the CRISPR protein. In some embodiments, porcine CD163 may be targeted by the CRISPR protein. In some embodiments, porcine CD163 may be knocked out through induction of a DSB or through insertions or deletions, for example targeting deletion or modification of exon 7, including one or more of those described above, or in other regions of the gene, for example deletion or modification of exon 5.

[00782] An edited pig and its progeny are also envisaged, for example a CD163 knock out pig. This may be for livestock, breeding or modelling purposes (i.e. a porcine model). Semen comprising the gene knock out is also provided.

[00783] CD163 is a member of the scavenger receptor cysteine-rich (SRCR) superfamily. Based on in vitro studies SRCR domain 5 of the protein is the domain responsible for unpackaging and release of the viral genome. As such, other members of the SRCR superfamily may also be targeted in order to assess resistance to other viruses. PRRSV is also a member of the mammalian arterivirus group, which also includes murine lactate dehydrogenase-elevating virus, simian hemorrhagic fever virus and equine arteritis virus. The arteriviruses share important pathogenesis properties, including macrophage

tropism and the capacity to cause both severe disease and persistent infection. Accordingly, arteriviruses, and in particular murine lactate dehydrogenase-elevating virus, simian hemorrhagic fever virus and equine arteritis virus, may be targeted, for example through porcine CD163 or homologues thereof in other species, and murine, simian and equine models and knockout also provided.

[00784] Indeed, this approach may be extended to viruses or bacteria that cause other livestock diseases that may be transmitted to humans, such as Swine Influenza Virus (SIV) strains which include influenza C and the subtypes of influenza A known as H1N1, H1N2, H2N1, H3N1, H3N2, and H2N3, as well as pneumonia, meningitis and oedema mentioned above.

[00785] In some embodiments, the AD-functionalized CRISPR system described herein can be used to genetically modify a pig genome to inactivate one or more porcine endogenous retrovirus (PERVs) loci to facilitate clinical application of porcine-to-human xenotransplantation. See Yang *et al.*, *Science* 350(6264):1101-1104 (2015), which is incorporated herein by reference in its entirey. In some embodiments, the AD-functionalized CRISPR system described herein can be used to produce a genetically modified pig that does not comprise any active porcine endogenous retrovirus (PERVs) locus.

Screens/Diagnostics/Treatments using CRISPR systems

[00786] Cancer

[00787] The methods and compositions of the invention can be used to identify cell states, components, and mechanisms associated with drug-tolerance and persistence of disease cells. Terai et al. (Cancer Research, 19-Dec-2017, doi: 10.1158/0008-5472.CAN-17-1904) reported a genome-wide CRISPR/Cas9 enhancer/suppressor screen in EGFR-dependent lung cancer PC9 cells treated with erlotinib + THZ1 (CDK7/12 inhibitor) combination therapy to identify multiple genes that enhanced erlotinib/THZ1 synergy, as well as components and pathways that suppress synergy. Wang et al. (Cell Rep. 2017 Feb 7;18(6):1543-1557. doi: 10.1016/j.celrep.2017.01.031.; Krall et al., Elife. 2017 Feb 1;6. pii: e18970. doi: 10.7554/eLife.18970) reported the use of genome-wide CRISPR loss-of-function screens to identify mediator of resistance to MAPK inhibitors. Donovan et al. (PLoS One. 2017 Jan 24;12(1):e0170445. doi: 10.1371/journal.pone.0170445. eCollection 2017) used a CRISPR-mediated approach to mutagenesis to identify novel gain-of-function and drug resistant alleles of the MAPK signaling pathway genes. Wang et al. (Cell. 2017

Feb 23;168(5):890-903.e15. doi: 10.1016/j.cell.2017.01.013. Epub 2017 Feb 2) used genome-wide CRISPR screens to identify gene networks and synthetic lethal interactions with oncogenic Ras. Chow et al. (Nat Neurosci. 2017 Oct;20(10):1329-1341. doi: 10.1038/nn.4620. Epub 2017 Aug 14) developed an adeno-associated virus-mediated, autochthonous genetic CRISPR screen in glioblastoma to identify functional suppressors in glioblastoma. Xue et al. (Nature. 2014 Oct 16;514(7522):380-4. doi: 10.1038/nature13589. Epub 2014 Aug 6) employed CRISPR-mediated direct mutation of cancer genes in the mouse liver.

[00788] Chen et al. (J Clin Invest. 2017 Dec 4. pii: 90793. doi: 10.1172/JCI90793. [Epub ahead of print]) used a CRISPR-based screen to identify MYCN-amplified neuroblastoma dependency on EZH2. Support testing of EZH2 inhibitors in patients with MYCN-amplified neuroblastoma.

[00789] Vijai et al. (Cancer Discov. 2016 Nov;6(11):1267-1275. Epub 2016 Sep 21) reported use of CRISPR to generate heterozygous mutations in the mammary epithelial cell line to assess risk for breast cancer.

[00790] Chakraborty et al. (Sci Transl Med. 2017 Jul 12;9(398). pii: eaal5272. doi: 10.1126/scitranslmed.aal5272) used a CRISPR-based screen to identify EZH1 as potential target to treat clear cell renal cell carcinoma

[00791] Metabolic disease

[00792] The methods and compositions of the invention provide advantages over conventional gene therapy methods in the treatment of inherited metabolic diseases of the liver, including but not limited to familial hypercholesterolemia, hemophilia, ornithine transcarbamylase deficiency, hereditary tyrosinemia type 1, and alpha-1 antitrypsin deficiency. See, Bryson et al., Yale J. Biol. Med. 90(4):553-566, 19-Dec-2017.

[00793] Bompada et al. (Int J Biochem Cell Biol. 2016 Dec;81(Pt A):82-91. doi: 10.1016/j.biocel.2016.10.022. Epub 2016 Oct 29) described the use of CRISPR to knockout histone acetyltransferase in pancreatic beta cells to demonstrate that histone acetylation serves as a key regulator of glucose-induced increase in TXNIP gene expression and thereby glucotoxicity-induced apoptosis.

[00794] Ocular

[00795] The invention provides efficient treatment of inherited and acquired ocular diseases of the retina. Holmgaard et al. (Mol. Ther. Nucleic Acids 9:89-99, 15-Dec-2017 doi: 10.1016/j.omtn.2017.08.016. Epub 2017 Sep 21) reported indel formation at high frequencies when SpCas9 was delivered by lentiviral vectors (LVs) encoding SpCas9

targeted to Vegfa and there was a significant reduction of of VEGFA in transduced cells. Duan et al. (J Biol Chem. 2016 Jul 29;291(31):16339-47. doi: 10.1074/jbc.M116.729467. Epub 2016 May 31) describe use of CRISPR to target MDM2 genomic locus in human primary retinal pigment epithelial cells

[00796] The methods and compositions of the instant invention are similarly applicable to treatment of ocular diseases, including age-related macular degeneration.

[00797] Huang et al. (Nat Commun. 2017 Jul 24;8(1):112. doi: 10.1038/s41467-017-00140-3 employed CRISPR to edit VEGFR2 to treat angiogenesis-associated diseases.

[00798] Hearing

[00799] Gao et al. (Nature. 2017 Dec 20. doi: 10.1038/nature25164. [Epub ahead of print]) reported genome editing using CRISPR-Cas9 to target Tmc1 gene in mice and reduce progressive hearing loss and deafness.

[00800] Muscle

[00801] Provenzano et al. (Mol Ther Nucleic Acids. 9:337-348. 15-Dec-2017;. doi: 10.1016/j.omtn.2017.10.006. Epub 2017 Oct 14) reported CRISPR/Cas9-mediated deletion of CTG expansions and permanent reversion to a normal phenotype in myogenic cells from myotonic dystrophy 1 patients. The methods and compositions of the instant invention are similarly applicable to nucleotide repeat disorders, not limited to CTG expansions. Tabebordbar et al. (2016 Jan 22;351(6271):407-411. doi: 10.1126/science.aad5177. Epub 2015 Dec 31) reports the use of CRISPR to edit the Dmd exon 23 locus to correct disruptive mutations in DMD. Tabebordbar shows that programmable CRISPR complexes can be delivered locally and systemically to terminally differentiated skeletal muscle fibers and cardiomyocytes, as well as muscle satellite cells, in neonatal and adult mice, where they mediate targeted gene modification, restore dystrophin expression and partially recover functional deficiencies of dystrophic muscle. See also Nelson et al., (Science. 2016 Jan 22;351(6271):403-7. doi: 10.1126/science.aad5143. Epub 2015 Dec 31).

[00802] Infectious disease

[00803] Sidik et al. (Cell. 2016 Sep 8;166(6):1423-1435.e12. doi: 10.1016/j.cell.2016.08.019. Epub 2016 Sep 2) and Patel et al. (Nature. 2017 Aug 31;548(7669):537-542. doi: 10.1038/nature23477. Epub 2017 Aug 7) describe a CRISPR screen in Toxoplasma and expansion of antiparasitic interventions.

[00804] There are several reports of genome-wide CRISPR screens to identify components and processes underlying host-pathogen interactions. Examples include Blondel et al. (Cell Host Microbe. 2016 Aug 10;20(2):226-37. doi:

10.1016/j.chom.2016.06.010. Epub 2016 Jul 21), Shapiro et al. (Nat Microbiol. 2018 Jan;3(1):73-82. doi: 10.1038/s41564-017-0043-0. Epub 2017 Oct 23) and Park et al. (Nat Genet. 2017 Feb;49(2):193-203. doi: 10.1038/ng.3741. Epub 2016 Dec 19).

[00805] Ma et al. (Cell Host Microbe. 2017 May 10;21(5):580-591.e7. doi: 10.1016/j.chom.2017.04.005) employed genome-wide CRISPR loss-of-function screens to identify viral transformation-driven synthetic lethal targets for therapeutic intervention.

[00806] Cardiovascular diseases

[00807] CRISPR systems can be used as to tool to identify genes or genetic variant associated with vascular disease. This is useful for identifying potential treatment or preventative targets. Xu et al. (Atherosclerosis, 2017 Sep. 21 pii: S0021-9150(17)31265-0. doi: 10.1016/j.atherosclerosis.2017.08.031. [Epub ahead of print]) reports the use of CRISPR to knockout the ANGPTL3 gene to confirm the role of ANGPTL3 in regulating plasma level of LDL-C. Gupta et al., (Cell. 2017 Jul 27;170(3):522-533.e15. doi: 10.1016/j.cell.2017.06.049) reports the use of CRISPR to edit stem cell-derived enthothelial cells to identify genetic variant associated with vascular diseases. Beaudoin et al., (Arterioscler Thromb Vasc Biol. 2015 Jun;35(6):1472-1479. doi: 10.1161/ATVBAHA.115.305534. Epub 2015 Apr 2), reports the use of CRISPR genome editing to disrupt binding of the transcription factors MEF2 at the locus. This sets the stage for exploring how PHACTR1 functions in the vascular endothelium influence coronary artery disease. Pashos et al. (Cell Stem Cell. 2017 Apr 6;20(4):558-570.e10. doi: 10.1016/j.stem.2017.03.017.) reports on using CRISPR technology to target pluripotent stem cells and hepatocyte-like cells to identify functional variants and lipid-functional genes.

[00808] In addition to being used as a tool for identifying targets, CRISPR systems can directly be used to treat or prevent cardiovascular diseases for known targets. Khera et al. (Nat Rev Genet. 2017 Jun;18(6):331-344. doi: 10.1038/nrg.2016.160. Epub 2017 Mar 13) described common variant association studies linking approximately 60 genetic loci to coronary risk used to facilitate a better understanding of causal risk factors, underlying biology development of new therapeutics. Khera explains, for example that inactivating mutations in PCSK9 decreased levels of circulating LDL cholesterol and reduced risk of CAD leading to intense interest in development of PCSK9 inhibitors. Further, antisense oligonucleotides designed to mimic protective mutations in APOC3 or LPA demonstrated a ~70% reduction in triglyceride levels and 80% reduction in circulating lipoprotein(a) levels, respectively. In addition, Wang et al., (Arterioscler Thromb Vasc Biol. 2016

May;36(5):783-6. doi: 10.1161/ATVBAHA.116.307227. Epub 2016 Mar 3) and Ding et al. (Circ Res. 2014 Aug 15;115(5):488-92. doi: 10.1161/CIRCRESAHA.115.304351. Epub 2014 Jun 10.) report the use of CRISPR to target the gene Pcsk9 for the prevention of cardiovascular disease.

[00809] The invention provides methods and compositions for investigating and treating neurological diseases and disorders. Nakayama et al., (Am J Hum Genet. 2015 May 7;96(5):709-19. doi: 10.1016/j.ajhg.2015.03.003. Epub 2015 Apr 9) report use of CRISPR to study the role of PYCR2 in human CNS development and to identify potential target for microcephaly and hypomyelination. Swiech et al. (Nat Biotechnol. 2015 Jan;33(1):102-6. doi: 10.1038/nbt.3055. Epub 2014 Oct 19) report use of CRISPR to target single (Mecp2) as well as multiple genes (Dnmt1, Dnmt3a and Dnmt3b) in the adult mouse brain *in vivo*. Shin et al. (Hum Mol Genet. 2016 Oct 15;25(20):4566-4576. doi: 10.1093/hmg/ddw286) describes the use of CRISPR to inactivate Huntingon's disease mutation. Platt et al. (Cell Rep. 2017 Apr 11;19(2):335-350. doi: 10.1016/j.celrep.2017.03.052) report use of CRISPR knockin mice to identify Chd8's role in autism spectrum disorder. Seo et al. (J Neurosci. 2017 Oct 11;37(41):9917-9924. doi: 10.1523/JNEUROSCI.0621-17.2017. Epub 2017 Sep 14) describe use of CRISPR to generate models of neurodegenerative disorders. Petersen et al. (Neuron. 2017 Dec 6;96(5):1003-1012.e7. doi: 10.1016/j.neuron.2017.10.008. Epub 2017 Nov 2) demonstrate CRISPR knockout of activin A receptor type I in oligodendrocyte progenitor cells to identify potential targets for diseases with remyelination failure. The methods and compositions of the instant invention are similarly applicable.

[00810] Other applications of CRISPR technology.

[00811] Renneville et al (Blood. 2015 Oct 15;126(16):1930-9. doi: 10.1182/blood-2015-06-649087. Epub 2015 Aug 28) report use of CRISPR to study the roles of EHMT1 and EMHT2 in fetal hemoglobin expression and to identify novel therapeutic target for SCD.

[00812] Tothova et al. (Cell Stem Cell. 2017 Oct 5;21(4):547-555.e8. doi: 10.1016/j.stem.2017.07.015) reported the use of CRISPR in hematopoietic stem and progenitor cells for generating models of human myeloid diseases.

[00813] Giani et al. (Cell Stem Cell. 2016 Jan 7;18(1):73-78. doi: 10.1016/j.stem.2015.09.015. Epub 2015 Oct 22) report that inactivation of SH2B3 by CRISPR/Cas9 genome editing in human pluripotent stem cells allowed enhanced erythroid cell expansion with preserved differentiation.

[00814] Wakabayashi et al. (Proc Natl Acad Sci U S A. 2016 Apr 19;113(16):4434-9. doi: 10.1073/pnas.1521754113. Epub 2016 Apr 4) employed CRISPR to gain insight into

GATA1 transcriptional activity and to investigate the pathogenicity of noncoding variants in human erythroid disorders.

[00815] Mandal et al. (Cell Stem Cell. 2014 Nov 6;15(5):643-52. doi: 10.1016/j.stem.2014.10.004. Epub 2014 Nov 6) describe CRISPR/Cas9 targeting of two clinically relevant genes, B2M and CCR5, in primary human CD4+ T cells and CD34+ hematopoietic stem and progenitor cells (HSPCs)

[00816] Polfus et al. (Am J Hum Genet. 2016 Sep 1;99(3):785. doi: 10.1016/j.ajhg.2016.08.002. Epub 2016 Sep 1) used CRISPR to edit hematopoietic cell lines and follow-up targeted knockdown experiments in primary human hematopoietic stem and progenitor cells and investigate the role of GFI1B variants in human hematopoiesis.

[00817] Najm et al. (Nat Biotechnol. 2017 Dec 18. doi: 10.1038/nbt.4048. [Epub ahead of print]) reports the use of CRISPR complex having a pair SaCas9 and SpCas9 to achieve dual targeting to generate high-complexity pooled dual-knockout libraries to identify synthetic lethal and buffering gene pairs across multiple cell types, including MAPK pathway genes and apoptotic genes.

[00818] Manguso et al. (Nature. 2017 Jul 27;547(7664):413-418. doi: 10.1038/nature23270. Epub 2017 Jul 19.) reports the use of CRISPR screens to identify and/or confirm new immunotherapy targets. See also Roland et al. (Proc Natl Acad Sci U S A. 2017 Jun 20;114(25):6581-6586. doi: 10.1073/pnas.1701263114. Epub 2017 Jun 12.); Erb et al. (Nature. 2017 Mar 9;543(7644):270-274. doi: 10.1038/nature21688. Epub 2017 Mar 1.); Hong et al., (Nat Commun. 2016 Jun 22;7:11987. doi: 10.1038/ncomms11987); Fei et al., (Proc Natl Acad Sci U S A. 2017 Jun 27;114(26):E5207-E5215. doi: 10.1073/pnas.1617467114. Epub 2017 Jun 13.); Zhang et al., (Cancer Discov. 2017 Sep 29. doi: 10.1158/2159-8290.CD-17-0532. [Epub ahead of print]).

[00819] Joung et al. (Nature. 2017 Aug 17;548(7667):343-346. doi: 10.1038/nature23451. Epub 2017 Aug 9.) reports the use of genome-wide screens to analyze long non-coding RNAs (lncRNA); see also Zhu et al., (Nat Biotechnol. 2016 Dec;34(12):1279-1286. doi: 10.1038/nbt.3715. Epub 2016 Oct 31); Sanjana et al., (Science. 2016 Sep 30;353(6307):1545-1549).

[00820] Barrow et al. (Mol Cell. 2016 Oct 6;64(1):163-175. doi: 10.1016/j.molcel.2016.08.023. Epub 2016 Sep 22.) reports the use of genome-wide CRISPR screens to search for therapeutic targets for mitochondrial diseases. See also Vafai et al., (PLoS One. 2016 Sep 13;11(9):e0162686. doi: 10.1371/journal.pone.0162686. eCollection 2016).

[00821] Guo et al. (Elife. 2017 Dec 5;6. pii: e29329. doi: 10.7554/eLife.29329) reports the use of CRISPR to target human chondrocytes to elucidate biological mechanisms for human growth.

[00822] Ramanan et al. (Sci Rep. 2015 Jun 2;5:10833. doi: 10.1038/srep10833) reports the use of CRISPR to target and cleave conserved regions in the HBV genome.

[00823] Therapeutic Targeting with AD-Functionalized CRISPR System

[00824] As will be apparent, it is envisaged that AD-functionalized CRISPR system can be used to target any polynucleotide sequence of interest. The invention provides a non-naturally occurring or engineered composition, or one or more polynucleotides encoding components of said composition, or vector or delivery systems comprising one or more polynucleotides encoding components of said composition for use in a modifying a target cell *in vivo*, *ex vivo* or *in vitro* and, may be conducted in a manner alters the cell such that once modified the progeny or cell line of the CRISPR modified cell retains the altered phenotype. The modified cells and progeny may be part of a multi-cellular organism such as a plant or animal with *ex vivo* or *in vivo* application of CRISPR system to desired cell types. The CRISPR invention may be a therapeutic method of treatment. The therapeutic method of treatment may comprise gene or genome editing, or gene therapy.

[00825] Adoptive Cell Therapies

[00826] The present invention also contemplates use of the AD-functionalized CRISPR system described herein to modify cells for adoptive therapies. Aspects of the invention accordingly involve the adoptive transfer of immune system cells, such as T cells, specific for selected antigens, such as tumor associated antigens (see Maus et al., 2014, Adoptive Immunotherapy for Cancer or Viruses, Annual Review of Immunology, Vol. 32: 189-225; Rosenberg and Restifo, 2015, Adoptive cell transfer as personalized immunotherapy for human cancer, Science Vol. 348 no. 6230 pp. 62-68; and, Restifo et al., 2015, Adoptive immunotherapy for cancer: harnessing the T cell response. Nat. Rev. Immunol. 12(4): 269-281; and Jenson and Riddell, 2014, Design and implementation of adoptive therapy with chimeric antigen receptor-modified T cells. Immunol Rev. 257(1): 127–144). Various strategies may for example be employed to genetically modify T cells by altering the specificity of the T cell receptor (TCR) for example by introducing new TCR α and β chains with selected peptide specificity (see U.S. Patent No. 8,697,854; PCT Patent Publications: WO2003020763, WO2004033685, WO2004044004, WO2005114215, WO2006000830, WO2008038002, WO2008039818, WO2004074322, WO2005113595, WO2006125962, WO2013166321, WO2013039889, WO2014018863, WO2014083173; U.S. Patent No. 8,088,379).

[00827] As an alternative to, or addition to, TCR modifications, chimeric antigen receptors (CARs) may be used in order to generate immunoresponsive cells, such as T cells, specific for selected targets, such as malignant cells, with a wide variety of receptor chimera constructs having been described (see U.S. Patent Nos. 5,843,728; 5,851,828; 5,912,170; 6,004,811; 6,284,240; 6,392,013; 6,410,014; 6,753,162; 8,211,422; and, PCT Publication WO9215322). Alternative CAR constructs may be characterized as belonging to successive generations. First-generation CARs typically consist of a single-chain variable fragment of an antibody specific for an antigen, for example comprising a VL linked to a VH of a specific antibody, linked by a flexible linker, for example by a CD8α hinge domain and a CD8α transmembrane domain, to the transmembrane and intracellular signaling domains of either CD3ζ or FcRγ (scFv-CD3ζ or scFv-FcRγ; see U.S. Patent No. 7,741,465; U.S. Patent No. 5,912,172; U.S. Patent No. 5,906,936). Second-generation CARs incorporate the intracellular domains of one or more costimulatory molecules, such as CD28, OX40 (CD134), or 4-1BB (CD137) within the endodomain (for example scFv-CD28/OX40/4-1BB-CD3ζ; see U.S. Patent Nos. 8,911,993; 8,916,381; 8,975,071; 9,101,584; 9,102,760; 9,102,761). Third-generation CARs include a combination of costimulatory endodomains, such a CD3ζ-chain, CD97, GDI la-CD18, CD2, ICOS, CD27, CD154, CDS, OX40, 4-1BB,

or CD28 signaling domains (for example scFv-CD28-4-1BB-CD3ζ or scFv-CD28-OX40-CD3ζ; see U.S. Patent No. 8,906,682; U.S. Patent No. 8,399,645; U.S. Pat. No. 5,686,281; PCT Publication No. WO2014134165; PCT Publication No. WO2012079000). Alternatively, costimulation may be orchestrated by expressing CARs in antigen-specific T cells, chosen so as to be activated and expanded following engagement of their native αβTCR, for example by antigen on professional antigen-presenting cells, with attendant costimulation. In addition, additional engineered receptors may be provided on the immunoresponsive cells, for example to improve targeting of a T-cell attack and/or minimize side effects.

[00828] Alternative techniques may be used to transform target immunoresponsive cells, such as protoplast fusion, lipofection, transfection or electroporation. A wide variety of vectors may be used, such as retroviral vectors, lentiviral vectors, adenoviral vectors, adeno-associated viral vectors, plasmids or transposons, such as a Sleeping Beauty transposon (see U.S. Patent Nos. 6,489,458; 7,148,203; 7,160,682; 7,985,739; 8,227,432), may be used to introduce CARs, for example using 2nd generation antigen-specific CARs signaling through CD3ζ and either CD28 or CD137. Viral vectors may for example include vectors based on HIV, SV40, EBV, HSV or BPV.

[00829] Cells that are targeted for transformation may for example include T cells, Natural Killer (NK) cells, cytotoxic T lymphocytes (CTL), regulatory T cells, human embryonic stem cells, tumor-infiltrating lymphocytes (TIL) or a pluripotent stem cell from which lymphoid cells may be differentiated. T cells expressing a desired CAR may for example be selected through co-culture with γ-irradiated activating and propagating cells (AaPC), which co-express the cancer antigen and co-stimulatory molecules. The engineered CAR T-cells may be expanded, for example by co-culture on AaPC in presence of soluble factors, such as IL-2 and IL-21. This expansion may for example be carried out so as to provide memory CAR+ T cells (which may for example be assayed by non-enzymatic digital array and/or multi-panel flow cytometry). In this way, CAR T cells may be provided that have specific cytotoxic activity against antigen-bearing tumors (optionally in conjunction with production of desired chemokines such as interferon-γ). CAR T cells of this kind may for example be used in animal models, for example to threat tumor xenografts.

[00830] Approaches such as the foregoing may be adapted to provide methods of treating and/or increasing survival of a subject having a disease, such as a neoplasia, for example by administering an effective amount of an immunoresponsive cell comprising an antigen

recognizing receptor that binds a selected antigen, wherein the binding activates the immunoreponsive cell, thereby treating or preventing the disease (such as a neoplasia, a pathogen infection, an autoimmune disorder, or an allogeneic transplant reaction). Dosing in CAR T cell therapies may for example involve administration of from 106 to 109 cells/kg, with or without a course of lymphodepletion, for example with cyclophosphamide.

[00831] In one embodiment, the treatment can be administrated into patients undergoing an immunosuppressive treatment. The cells or population of cells, may be made resistant to at least one immunosuppressive agent due to the inactivation of a gene encoding a receptor for such immunosuppressive agent. Not being bound by a theory, the immunosuppressive treatment should help the selection and expansion of the immunoresponsive or T cells according to the invention within the patient.

[00832] The administration of the cells or population of cells according to the present invention may be carried out in any convenient manner, including by aerosol inhalation, injection, ingestion, transfusion, implantation or transplantation. The cells or population of cells may be administered to a patient subcutaneously, intradermally, intratumorally, intranodally, intramedullary, intramuscularly, by intravenous or intralymphatic injection, or intraperitoneally. In one embodiment, the cell compositions of the present invention are preferably administered by intravenous injection.

[00833] The administration of the cells or population of cells can consist of the administration of 10^4- 10^9 cells per kg body weight, preferably 10^5 to 10^6 cells/kg body weight including all integer values of cell numbers within those ranges. Dosing in CAR T cell therapies may for example involve administration of from 10^6 to 10^9 cells/kg, with or without a course of lymphodepletion, for example with cyclophosphamide. The cells or population of cells can be administrated in one or more doses. In another embodiment, the effective amount of cells are administrated as a single dose. In another embodiment, the effective amount of cells are administrated as more than one dose over a period time. Timing of administration is within the judgment of managing physician and depends on the clinical condition of the patient. The cells or population of cells may be obtained from any source, such as a blood bank or a donor. While individual needs vary, determination of optimal ranges of effective amounts of a given cell type for a particular disease or conditions are within the skill of one in the art. An effective amount means an amount which provides a therapeutic or prophylactic benefit. The dosage administrated will be dependent upon the age, health and weight of the recipient, kind of concurrent treatment, if any, frequency of treatment and the nature of the effect desired.

[00834] In another embodiment, the effective amount of cells or composition comprising those cells are administrated parenterally. The administration can be an intravenous administration. The administration can be directly done by injection within a tumor.

[00835] To guard against possible adverse reactions, engineered immunoresponsive cells may be equipped with a transgenic safety switch, in the form of a transgene that renders the cells vulnerable to exposure to a specific signal. For example, the herpes simplex viral thymidine kinase (TK) gene may be used in this way, for example by introduction into allogeneic T lymphocytes used as donor lymphocyte infusions following stem cell transplantation (Greco, et al., Improving the safety of cell therapy with the TK-suicide gene. Front. Pharmacol. 2015; 6: 95). In such cells, administration of a nucleoside prodrug such as ganciclovir or acyclovir causes cell death. Alternative safety switch constructs include inducible caspase 9, for example triggered by administration of a small-molecule dimerizer that brings together two nonfunctional icasp9 molecules to form the active enzyme. A wide variety of alternative approaches to implementing cellular proliferation controls have been described (see U.S. Patent Publication No. 20130071414; PCT Patent Publication WO2011146862; PCT Patent Publication WO2014011987; PCT Patent Publication WO2013040371; Zhou et al. BLOOD, 2014, 123/25:3895 – 3905; Di Stasi et al., The New England Journal of Medicine 2011; 365:1673-1683; Sadelain M, The New England Journal of Medicine 2011; 365:1735-173; Ramos et al., Stem Cells 28(6):1107-15 (2010)).

[00836] In a further refinement of adoptive therapies, genome editing with a AD-functionalized CRISPR-Cas system as described herein may be used to tailor immunoresponsive cells to alternative implementations, for example providing edited CAR T cells (see Poirot et al., 2015, Multiplex genome edited T-cell manufacturing platform for "off-the-shelf" adoptive T-cell immunotherapies, Cancer Res 75 (18): 3853). For example, immunoresponsive cells may be edited to delete expression of some or all of the class of HLA type II and/or type I molecules, or to knockout selected genes that may inhibit the desired immune response, such as the PD1 gene.

[00837] Cells may be edited using a AD-functionalized CRISPR system as described herein. AD-functionalized CRISPR systems may be delivered to an immune cell by any method described herein. In preferred embodiments, cells are edited *ex vivo* and transferred to a subject in need thereof. Immunoresponsive cells, CAR-T cells or any cells used for adoptive cell transfer may be edited. Editing may be performed to eliminate potential alloreactive T-cell receptors (TCR), disrupt the target of a chemotherapeutic agent, block an immune checkpoint, activate a T cell, and/or increase the differentiation and/or proliferation

of functionally exhausted or dysfunctional CD8+ T-cells (see PCT Patent Publications: WO2013176915, WO2014059173, WO2014172606, WO2014184744, and WO2014191128). Editing may result in inactivation of a gene.

[00838] T cell receptors (TCR) are cell surface receptors that participate in the activation of T cells in response to the presentation of antigen. The TCR is generally made from two chains, α and β, which assemble to form a heterodimer and associates with the CD3-transducing subunits to form the T cell receptor complex present on the cell surface. Each α and β chain of the TCR consists of an immunoglobulin-like N-terminal variable (V) and constant (C) region, a hydrophobic transmembrane domain, and a short cytoplasmic region. As for immunoglobulin molecules, the variable region of the α and β chains are generated by V(D)J recombination, creating a large diversity of antigen specificities within the population of T cells. However, in contrast to immunoglobulins that recognize intact antigen, T cells are activated by processed peptide fragments in association with an MHC molecule, introducing an extra dimension to antigen recognition by T cells, known as MHC restriction. Recognition of MHC disparities between the donor and recipient through the T cell receptor leads to T cell proliferation and the potential development of graft versus host disease (GVHD). The inactivation of TCRα or TCRβ can result in the elimination of the TCR from the surface of T cells preventing recognition of alloantigen and thus GVHD. However, TCR disruption generally results in the elimination of the CD3 signaling component and alters the means of further T cell expansion.

[00839] Allogeneic cells are rapidly rejected by the host immune system. It has been demonstrated that, allogeneic leukocytes present in non-irradiated blood products will persist for no more than 5 to 6 days (Boni, Muranski et al. 2008 Blood 1;112(12):4746-54). Thus, to prevent rejection of allogeneic cells, the host's immune system usually has to be suppressed to some extent. However, in the case of adoptive cell transfer the use of immunosuppressive drugs also have a detrimental effect on the introduced therapeutic T cells. Therefore, to effectively use an adoptive immunotherapy approach in these conditions, the introduced cells would need to be resistant to the immunosuppressive treatment. Thus, in a particular embodiment, the present invention further comprises a step of modifying T cells to make them resistant to an immunosuppressive agent, preferably by inactivating at least one gene encoding a target for an immunosuppressive agent. An immunosuppressive agent is an agent that suppresses immune function by one of several mechanisms of action. An immunosuppressive agent can be, but is not limited to a calcineurin inhibitor, a target of rapamycin, an interleukin-2 receptor α-chain blocker, an

inhibitor of inosine monophosphate dehydrogenase, an inhibitor of dihydrofolic acid reductase, a corticosteroid or an immunosuppressive antimetabolite. The present invention allows conferring immunosuppressive resistance to T cells for immunotherapy by inactivating the target of the immunosuppressive agent in T cells. As non-limiting examples, targets for an immunosuppressive agent can be a receptor for an immunosuppressive agent such as: CD52, glucocorticoid receptor (GR), a FKBP family gene member and a cyclophilin family gene member.

[00840] Immune checkpoints are inhibitory pathways that slow down or stop immune reactions and prevent excessive tissue damage from uncontrolled activity of immune cells. In certain embodiments, the immune checkpoint targeted is the programmed death-1 (PD-1 or CD279) gene (*PDCD1*). In other embodiments, the immune checkpoint targeted is cytotoxic T-lymphocyte-associated antigen (CTLA-4). In additional embodiments, the immune checkpoint targeted is another member of the CD28 and CTLA4 Ig superfamily such as BTLA, LAG3, ICOS, PDL1 or KIR. In further additional embodiments, the immune checkpoint targeted is a member of the TNFR superfamily such as CD40, OX40, CD137, GITR, CD27 or TIM-3.

[00841] Additional immune checkpoints include Src homology 2 domain-containing protein tyrosine phosphatase 1 (SHP-1) (Watson HA, et al., SHP-1: the next checkpoint target for cancer immunotherapy? Biochem Soc Trans. 2016 Apr 15;44(2):356-62). SHP-1 is a widely expressed inhibitory protein tyrosine phosphatase (PTP). In T-cells, it is a negative regulator of antigen-dependent activation and proliferation. It is a cytosolic protein, and therefore not amenable to antibody-mediated therapies, but its role in activation and proliferation makes it an attractive target for genetic manipulation in adoptive transfer strategies, such as chimeric antigen receptor (CAR) T cells. Immune checkpoints may also include T cell immunoreceptor with Ig and ITIM domains (TIGIT/Vstm3/WUCAM/VSIG9) and VISTA (Le Mercier I, et al., (2015) Beyond CTLA-4 and PD-1, the generation Z of negative checkpoint regulators. Front. Immunol. 6:418).

[00842] WO2014172606 relates to the use of MT1 and/or MT1 inhibitors to increase proliferation and/or activity of exhausted CD8+ T-cells and to decrease CD8+ T-cell exhaustion (e.g., decrease functionally exhausted or unresponsive CD8+ immune cells). In certain embodiments, metallothioneins are targeted by gene editing in adoptively transferred T cells.

[00843] In certain embodiments, targets of gene editing may be at least one targeted locus involved in the expression of an immune checkpoint protein. Such targets may

include, but are not limited to CTLA4, PPP2CA, PPP2CB, PTPN6, PTPN22, PDCD1, ICOS (CD278), PDL1, KIR, LAG3, HAVCR2, BTLA, CD160, TIGIT, CD96, CRTAM, LAIR1, SIGLEC7, SIGLEC9, CD244 (2B4), TNFRSF10B, TNFRSF10A, CASP8, CASP10, CASP3, CASP6, CASP7, FADD, FAS, TGFBRII, TGFRBRI, SMAD2, SMAD3, SMAD4, SMAD10, SKI, SKIL, TGIF1, IL10RA, IL10RB, HMOX2, IL6R, IL6ST, EIF2AK4, CSK, PAG1, SIT1, FOXP3, PRDM1, BATF, VISTA, GUCY1A2, GUCY1A3, GUCY1B2, GUCY1B3, MT1, MT2, CD40, OX40, CD137, GITR, CD27, SHP-1 or TIM-3. In preferred embodiments, the gene locus involved in the expression of PD-1 or CTLA-4 genes is targeted. In other preferred embodiments, combinations of genes are targeted, such as but not limited to PD-1 and TIGIT.

[00844] In other embodiments, at least two genes are edited. Pairs of genes may include, but are not limited to PD1 and TCRα, PD1 and TCRβ, CTLA-4 and TCRα, CTLA-4 and TCRβ, LAG3 and TCRα, LAG3 and TCRβ, Tim3 and TCRα, Tim3 and TCRβ, BTLA and TCRα, BTLA and TCRβ, BY55 and TCRα, BY55 and TCRβ, TIGIT and TCRα, TIGIT and TCRβ, B7H5 and TCRα, B7H5 and TCRβ, LAIR1 and TCRα, LAIR1 and TCRβ, SIGLEC10 and TCRα, SIGLEC10 and TCRβ, 2B4 and TCRα, 2B4 and TCRβ.

[00845] Whether prior to or after genetic modification of the T cells, the T cells can be activated and expanded generally using methods as described, for example, in U.S. Patents 6,352,694; 6,534,055; 6,905,680; 5,858,358; 6,887,466; 6,905,681; 7,144,575; 7,232,566; 7,175,843; 5,883,223; 6,905,874; 6,797,514; 6,867,041; and 7,572,631. T cells can be expanded *in vitro* or *in vivo*.

[00846] The practice of the present invention employs techniques known in the field of immunology, biochemistry, chemistry, molecular biology, microbiology, cell biology, genomics and recombinant DNA, which are within the skill of the art. See MOLECULAR CLONING: A LABORATORY MANUAL, 2nd edition (1989) (Sambrook, Fritsch and Maniatis); MOLECULAR CLONING: A LABORATORY MANUAL, 4th edition (2012) (Green and Sambrook); CURRENT PROTOCOLS IN MOLECULAR BIOLOGY (1987) (F. M. Ausubel, et al. eds.); the series METHODS IN ENZYMOLOGY (Academic Press, Inc.); PCR 2: A PRACTICAL APPROACH (1995) (M.J. MacPherson, B.D. Hames and G.R. Taylor eds.); ANTIBODIES, A LABORATORY MANUAL (1988) (Harlow and Lane, eds.); ANTIBODIES A LABORATORY MANUAL, 2nd edition (2013) (E.A. Greenfield ed.); and ANIMAL CELL CULTURE (1987) (R.I. Freshney, ed.).

[00847] **Correction of Disease-Associated Mutations and Pathogenic SNPs**

[00848] In one aspect, the invention described herein provides methods for modifying an adenosine residue at a target locus with the aim of remedying and/or preventing a diseased condition that is or is likely to be caused by a G-to-A or C-to-T point mutation or a pathogenic single nucleotide polymorphism (SNP).

[00849] **Diseases Affecting the Brain and Central Nervous System**

[00850] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various diseases affecting the brain and central nervous system are reported in the ClinVar database and disclosed in Table A, including but not limited to Alzheimer's Disease, Parkinson's Disease, Autism, Amyotrophyic lateral sclerosis (ALS), Schizophrenia, Adrenoleukodystrophy, Aicardi Goutieres syndrome, Fabry disease, Lesch-Nyhan syndrome, and Menkes Disease. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00851] Alzheimer's Disease

[00852] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Alzheimer's Disease. In some embodiments, the pathogenic mutations/SNPs are present in at least one gene selected from PSEN1, PSEN2, and APP, including at least the followings:

NM_000021.3(PSEN1):c.796G>A (p.Gly266Ser)

NM_000484.3(APP):c.2017G>A (p.Ala673Thr)

NM_000484.3(APP):c.2149G>A (p.Val717Ile)

NM_000484.3(APP):c.2137G>A (p.Ala713Thr)

NM_000484.3(APP):c.2143G>A (p.Val715Met)

NM_000484.3(APP):c.2141C>T (p.Thr714Ile)

NM_000021.3(PSEN1):c.438G>A (p.Met146Ile)

NM_000021.3(PSEN1):c.1229G>A (p.Cys410Tyr)

NM_000021.3(PSEN1):c.487C>T (p.His163Tyr)

NM_000021.3(PSEN1):c.799C>T (p.Pro267Ser)

NM_000021.3(PSEN1):c.236C>T (p.Ala79Val)

NM_000021.3(PSEN1):c.509C>T (p.Ser170Phe)

NM_000447.2(PSEN2):c.1289C>T (p.Thr430Met)

NM_000447.2(PSEN2):c.717G>A (p.Met239Ile)

NM_000447.2(PSEN2):c.254C>T (p.Ala85Val)

NM_000021.3(PSEN1):c.806G>A (p.Arg269His)

NM_000484.3(APP):c.2018C>T (p.Ala673Val).

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Alzheimer's Disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from PSEN1, PSEN2, and APP, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00853] Parkinson's Disease

[00854] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Parkinson's Disease. In some embodiments, In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from SNCA, PLA2G6, FBXO7, VPS35, EIF4G1, DNAJC6, PRKN, SYNJ1, CHCHD2, PINK1, PARK7, LRRK2, ATP13A2, and GBA, including at least the followings:

NM_000345.3(SNCA):c.157G>A (p.Ala53Thr)

NM_000345.3(SNCA):c.152G>A (p.Gly51Asp)

NM_003560.3(PLA2G6):c.2222G>A (p.Arg741Gln)

NM_003560.3(PLA2G6):c.2239C>T (p.Arg747Trp)

NM_003560.3(PLA2G6):c.1904G>A (p.Arg635Gln)

NM_003560.3(PLA2G6):c.1354C>T (p.Gln452Ter)

NM_012179.3(FBXO7):c.1492C>T (p.Arg498Ter)

NM_012179.3(FBXO7):c.65C>T (p.Thr22Met)

NM_018206.5(VPS35):c.1858G>A (p.Asp620Asn)

NM_198241.2(EIF4G1):c.3614G>A (p.Arg1205His)

NM_198241.2(EIF4G1):c.1505C>T (p.Ala502Val)

NM_001256865.1(DNAJC6):c.2200C>T (p.Gln734Ter)

NM_001256865.1(DNAJC6):c.2326C>T (p.Gln776Ter)

NM_004562.2(PRKN):c.931C>T (p.Gln311Ter)

NM_004562.2(PRKN):c.1358G>A (p.Trp453Ter)

NM_004562.2(PRKN):c.635G>A (p.Cys212Tyr)

NM_203446.2(SYNJ1):c.773G>A (p.Arg258Gln)

NM_001320327.1(CHCHD2):c.182C>T (p.Thr61Ile)

NM_001320327.1(CHCHD2):c.434G>A (p.Arg145Gln)

NM_001320327.1(CHCHD2):c.300+5G>A

NM_032409.2(PINK1):c.926G>A (p.Gly309Asp)
NM_032409.2(PINK1):c.1311G>A (p.Trp437Ter)
NM_032409.2(PINK1):c.736C>T (p.Arg246Ter)
NM_032409.2(PINK1):c.836G>A (p.Arg279His)
NM_032409.2(PINK1):c.938C>T (p.Thr313Met)
NM_032409.2(PINK1):c.1366C>T (p.Gln456Ter)
NM_007262.4(PARK7):c.78G>A (p.Met26Ile)
NM_198578.3(LRRK2):c.4321C>T (p.Arg1441Cys)
NM_198578.3(LRRK2):c.4322G>A (p.Arg1441His)
NM_198578.3(LRRK2):c.1256C>T (p.Ala419Val)
NM_198578.3(LRRK2):c.6055G>A (p.Gly2019Ser)
NM_022089.3(ATP13A2):c.1306+5G>A
NM_022089.3(ATP13A2):c.2629G>A (p.Gly877Arg)
NM_022089.3(ATP13A2):c.490C>T (p.Arg164Trp)
NM_001005741.2(GBA):c.1444G>A (p.Asp482Asn)
m.15950G>A.

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Parkinson's Disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs in at least one gene selected from SNCA, PLA2G6, FBXO7, VPS35, EIF4G1, DNAJC6, PRKN, SYNJ1, CHCHD2, PINK1, PARK7, LRRK2, ATP13A2, and GBA, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00855] Autism

[00856] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Autism. In some embodiments, the pathogenic mutations/SNPs are present in at least one gene selected from MECP2, NLGN3, SLC9A9, EHMT1, CHD8, NLGN4X , GSPT2, and PTEN, including at least the followings:

NM_001110792.1(MECP2):c.916C>T (p.Arg306Ter)
NM_004992.3(MECP2):c.473C>T (p.Thr158Met)
NM_018977.3(NLGN3):c.1351C>T (p.Arg451Cys)
NM_173653.3(SLC9A9):c.1267C>T (p.Arg423Ter)
NM_024757.4(EHMT1):c.3413G>A (p.Trp1138Ter)
NM_020920.3(CHD8):c.2875C>T (p.Gln959Ter)

NM_020920.3(CHD8):c.3172C>T (p.Arg1058Ter)

NM_181332.2(NLGN4X):c.301C>T (p.Arg101Ter)

NM_018094.4(GSPT2):c.1021G>A (p.Val341Ile)

NM_000314.6(PTEN):c.392C>T (p.Thr131Ile)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Autism by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from MECP2, NLGN3, SLC9A9, EHMT1, CHD8, NLGN4X , GSPT2, and PTEN, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00857] Amyotrophyic lateral sclerosis (ALS)

[00858] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with ALS. In some embodiments, the pathogenic mutations/SNPs are present in at least one gene selected from SOD1, VCP, UBQLN2, ERBB4, HNRNPA1, TUBA4A, SOD1, TARDBP, FIG4, OPTN, SETX, SPG11, FUS, VAPB, ANG, CHCHD10, SQSTM1, and TBK1, including at least the followings:

NM_000454.4(SOD1):c.289G>A (p.Asp97Asn)

NM_007126.3(VCP):c.1774G>A (p.Asp592Asn)

NM_007126.3(VCP):c.464G>A (p.Arg155His)

NM_007126.3(VCP):c.572G>A (p.Arg191Gln)

NM_013444.3(UBQLN2):c.1489C>T (p.Pro497Ser)

NM_013444.3(UBQLN2):c.1525C>T (p.Pro509Ser)

NM_013444.3(UBQLN2):c.1573C>T (p.Pro525Ser)

NM_013444.3(UBQLN2):c.1490C>T (p.Pro497Leu)

NM_005235.2(ERBB4):c.2780G>A (p.Arg927Gln)

NM_005235.2(ERBB4):c.3823C>T (p.Arg1275Trp)

NM_031157.3(HNRNPA1):c.940G>A (p.Asp314Asn)

NM_006000.2(TUBA4A):c.643C>T (p.Arg215Cys)

NM_006000.2(TUBA4A):c.958C>T (p.Arg320Cys)

NM_006000.2(TUBA4A):c.959G>A (p.Arg320His)

NM_006000.2(TUBA4A):c.1220G>A (p.Trp407Ter)

NM_006000.2(TUBA4A):c.1147G>A (p.Ala383Thr)

NM_000454.4(SOD1):c.112G>A (p.Gly38Arg)

NM_000454.4(SOD1):c.124G>A (p.Gly42Ser)

NM_000454.4(SOD1):c.125G>A (p.Gly42Asp)

NM_000454.4(SOD1):c.14C>T (p.Ala5Val)

NM_000454.4(SOD1):c.13G>A (p.Ala5Thr)

NM_000454.4(SOD1):c.436G>A (p.Ala146Thr)

NM_000454.4(SOD1):c.64G>A (p.Glu22Lys)

NM_000454.4(SOD1):c.404G>A (p.Ser135Asn)

NM_000454.4(SOD1):c.49G>A (p.Gly17Ser)

NM_000454.4(SOD1):c.217G>A (p.Gly73Ser)

NM_007375.3(TARDBP):c.892G>A (p.Gly298Ser)

NM_007375.3(TARDBP):c.943G>A (p.Ala315Thr)

NM_007375.3(TARDBP):c.883G>A (p.Gly295Ser)

NM_007375.3(TARDBP):c.*697G>A

NM_007375.3(TARDBP):c.1144G>A (p.Ala382Thr)

NM_007375.3(TARDBP):c.859G>A (p.Gly287Ser)

NM_014845.5(FIG4):c.547C>T (p.Arg183Ter)

NM_001008211.1(OPTN):c.1192C>T (p.Gln398Ter)

NM_015046.5(SETX):c.6407G>A (p.Arg2136His)

NM_015046.5(SETX):c.8C>T (p.Thr3Ile)

NM_025137.3(SPG11):c.118C>T (p.Gln40Ter)

NM_025137.3(SPG11):c.267G>A (p.Trp89Ter)

NM_025137.3(SPG11):c.5974C>T (p.Arg1992Ter)

NM_004960.3(FUS):c.1553G>A (p.Arg518Lys)

NM_004960.3(FUS):c.1561C>T (p.Arg521Cys)

NM_004960.3(FUS):c.1562G>A (p.Arg521His)

NM_004960.3(FUS):c.1520G>A (p.Gly507Asp)

NM_004960.3(FUS):c.1483C>T (p.Arg495Ter)

NM_004960.3(FUS):c.616G>A (p.Gly206Ser)

NM_004960.3(FUS):c.646C>T (p.Arg216Cys)

NM_004738.4(VAPB):c.166C>T (p.Pro56Ser)

NM_004738.4(VAPB):c.137C>T (p.Thr46Ile)

NM_001145.4(ANG):c.164G>A (p.Arg55Lys)

NM_001145.4(ANG):c.155G>A (p.Ser52Asn)

NM_001145.4(ANG):c.407C>T (p.Pro136Leu)

NM_001145.4(ANG):c.409G>A (p.Val137Ile)

NM_001301339.1(CHCHD10):c.239C>T (p.Pro80Leu)

NM_001301339.1(CHCHD10):c.176C>T (p.Ser59Leu)

NM_001142298.1(SQSTM1):c.-47-1924C>T

NM_003900.4(SQSTM1):c.1160C>T (p.Pro387Leu)

NM_003900.4(SQSTM1):c.1175C>T (p.Pro392Leu)

NM_013254.3(TBK1):c.1340+1G>A

NM_013254.3(TBK1):c.2086G>A (p.Glu696Lys)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing ALS by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from SOD1, VCP, UBQLN2, ERBB4, HNRNPA1, TUBA4A, SOD1, TARDBP, FIG4, OPTN, SETX, SPG11, FUS, VAPB, ANG, CHCHD10, SQSTM1, and TBK1,, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00859] Schizophrenia

[00860] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Schizophrenia. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from PRODH, SETD1A, and SHANK3, including at least the followings:

NM_016335.4(PRODH):c.1292G>A (p.Arg431His)

NM_016335.4(PRODH):c.1397C>T (p.Thr466Met)

NM_014712.2(SETD1A):c.2209C>T (p.Gln737Ter)

NM_033517.1(SHANK3):c.3349C>T (p.Arg1117Ter)

NM_033517.1(SHANK3):c.1606C>T (p.Arg536Trp)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Schizophrenia by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from PRODH, SETD1A, and SHANK3, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00861] Adrenoleukodystrophy

[00862] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated

with Adrenoleukodystrophy. In some embodiment, the pathogenic mutations/SNPs are present in at least the ABCD1 gene, including at least the followings:

NM_000033.3(ABCD1):c.421G>A (p.Ala141Thr)

NM_000033.3(ABCD1):c.796G>A (p.Gly266Arg)

NM_000033.3(ABCD1):c.1252C>T (p.Arg418Trp)

NM_000033.3(ABCD1):c.1552C>T (p.Arg518Trp)

NM_000033.3(ABCD1):c.1850G>A (p.Arg617His)

NM_000033.3(ABCD1):c.1396C>T (p.Gln466Ter)

NM_000033.3(ABCD1):c.1553G>A (p.Arg518Gln)

NM_000033.3(ABCD1):c.1679C>T (p.Pro560Leu)

NM_000033.3(ABCD1):c.1771C>T (p.Arg591Trp)

NM_000033.3(ABCD1):c.1802G>A (p.Trp601Ter)

NM_000033.3(ABCD1):c.346G>A (p.Gly116Arg)

NM_000033.3(ABCD1):c.406C>T (p.Gln136Ter)

NM_000033.3(ABCD1):c.1661G>A (p.Arg554His)

NM_000033.3(ABCD1):c.1825G>A (p.Glu609Lys)

NM_000033.3(ABCD1):c.1288C>T (p.Gln430Ter)

NM_000033.3(ABCD1):c.1781-1G>A

NM_000033.3(ABCD1):c.529C>T (p.Gln177Ter)

NM_000033.3(ABCD1):c.1866-10G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Adrenoleukodystrophy by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least the ABCD1 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00863] Aicardi Goutieres syndrome

[00864] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Aicardi Goutieres syndrome. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from TREX1, RNASEH2C, ADAR, and IFIH1, including at least the followings:

NM_016381.5(TREX1):c.794G>A (p.Trp265Ter)

NM_033629.4(TREX1):c.52G>A (p.Asp18Asn)

NM_033629.4(TREX1):c.490C>T (p.Arg164Ter)

NM_032193.3(RNASEH2C):c.205C>T (p.Arg69Trp)

NM_001111.4(ADAR):c.3019G>A (p.Gly1007Arg)

NM_022168.3(IFIH1):c.2336G>A (p.Arg779His)

NM_022168.3(IFIH1):c.2335C>T (p.Arg779Cys)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Aicardi Goutieres syndrome by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from TREX1, RNASEH2C, ADAR, and IFIH1, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00865] Fabry disease

[00866] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Fabry disease. In some embodiment, the pathogenic mutations/SNPs are present in at least the GLA gene, including at least the followings:

NM_000169.2(GLA):c.1024C>T (p.Arg342Ter)

NM_000169.2(GLA):c.1066C>T (p.Arg356Trp)

NM_000169.2(GLA):c.1025G>A (p.Arg342Gln)

NM_000169.2(GLA):c.281G>A (p.Cys94Tyr)

NM_000169.2(GLA):c.677G>A (p.Trp226Ter)

NM_000169.2(GLA):c.734G>A (p.Trp245Ter)

NM_000169.2(GLA):c.748C>T (p.Gln250Ter)

NM_000169.2(GLA):c.658C>T (p.Arg220Ter)

NM_000169.2(GLA):c.730G>A (p.Asp244Asn)

NM_000169.2(GLA):c.369+1G>A

NM_000169.2(GLA):c.335G>A (p.Arg112His)

NM_000169.2(GLA):c.485G>A (p.Trp162Ter)

NM_000169.2(GLA):c.661C>T (p.Gln221Ter)

NM_000169.2(GLA):c.916C>T (p.Gln306Ter)

NM_000169.2(GLA):c.1072G>A (p.Glu358Lys)

NM_000169.2(GLA):c.1087C>T (p.Arg363Cys)

NM_000169.2(GLA):c.1088G>A (p.Arg363His)

NM_000169.2(GLA):c.605G>A (p.Cys202Tyr)

NM_000169.2(GLA):c.830G>A (p.Trp277Ter)

NM_000169.2(GLA):c.979C>T (p.Gln327Ter)

NM_000169.2(GLA):c.422C>T (p.Thr141Ile)

NM_000169.2(GLA):c.285G>A (p.Trp95Ter)

NM_000169.2(GLA):c.735G>A (p.Trp245Ter)

NM_000169.2(GLA):c.639+919G>A

NM_000169.2(GLA):c.680G>A (p.Arg227Gln)

NM_000169.2(GLA):c.679C>T (p.Arg227Ter)

NM_000169.2(GLA):c.242G>A (p.Trp81Ter)

NM_000169.2(GLA):c.901C>T (p.Arg301Ter)

NM_000169.2(GLA):c.974G>A (p.Gly325Asp)

NM_000169.2(GLA):c.847C>T (p.Gln283Ter)

NM_000169.2(GLA):c.469C>T (p.Gln157Ter)

NM_000169.2(GLA):c.1118G>A (p.Gly373Asp)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Fabry disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least the GLA gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00867] Lesch-Nyhan syndrome

[00868] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Lesch-Nyhan syndrome. In some embodiment, the pathogenic mutations/SNPs are present in at least the HPRT1 gene, including at least the followings:

NM_000194.2(HPRT1):c.151C>T (p.Arg51Ter)

NM_000194.2(HPRT1):c.384+1G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Lesch-Nyhan syndrome by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least the HPRT1 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00869] Menkes Disease

[00870] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated

with Menkes Disease. In some embodiment, the pathogenic mutations/SNPs are present in at least the ATP7A gene, including at least the followings:

NM_000052.6(ATP7A):c.601C>T (p.Arg201Ter)

NM_000052.6(ATP7A):c.2938C>T (p.Arg980Ter)

NM_000052.6(ATP7A):c.3056G>A (p.Gly1019Asp)

NM_000052.6(ATP7A):c.598C>T (p.Gln200Ter)

NM_000052.6(ATP7A):c.1225C>T (p.Arg409Ter)

NM_000052.6(ATP7A):c.1544-1G>A

NM_000052.6(ATP7A):c.1639C>T (p.Arg547Ter)

NM_000052.6(ATP7A):c.1933C>T (p.Arg645Ter)

NM_000052.6(ATP7A):c.1946+5G>A

NM_000052.6(ATP7A):c.1950G>A (p.Trp650Ter)

NM_000052.6(ATP7A):c.2179G>A (p.Gly727Arg)

NM_000052.6(ATP7A):c.2187G>A (p.Trp729Ter)

NM_000052.6(ATP7A):c.2383C>T (p.Arg795Ter)

NM_000052.6(ATP7A):c.2499-1G>A

NM_000052.6(ATP7A):c.2555C>T (p.Pro852Leu)

NM_000052.6(ATP7A):c.2956C>T (p.Arg986Ter)

NM_000052.6(ATP7A):c.3112-1G>A

NM_000052.6(ATP7A):c.3466C>T (p.Gln1156Ter)

NM_000052.6(ATP7A):c.3502C>T (p.Gln1168Ter)

NM_000052.6(ATP7A):c.3764G>A (p.Gly1255Glu)

NM_000052.6(ATP7A):c.3943G>A (p.Gly1315Arg)

NM_000052.6(ATP7A):c.4123+1G>A

NM_000052.6(ATP7A):c.4226+5G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Menkes Disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least the ATP7A gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00871] Eye Diseases

[00872] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various eye diseases are reported in the ClinVar database and disclosed in Table A, including but not limited to Stargardt Disease, Bardet-Biedl Syndrome, Cone-rod dystrophy, Congenital

Stationary Night Blindness, Usher Syndrome, Leber Congenital Amaurosis, Retinitis Pigmentosa, and Achromatopsia. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00873] Stargardt Disease

[00874] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Stargardt Disease. In some embodiment, the pathogenic mutations/SNPs are present in the ABCA4 gene, including at least the followings:

NM_000350.2(ABCA4):c.4429C>T (p.Gln1477Ter)

NM_000350.2(ABCA4):c.6647C>T (p.Ala2216Val)

NM_000350.2(ABCA4):c.5312+1G>A

NM_000350.2(ABCA4):c.5189G>A (p.Trp1730Ter)

NM_000350.2(ABCA4):c.4352+1G>A

NM_000350.2(ABCA4):c.4253+5G>A

NM_000350.2(ABCA4):c.3871C>T (p.Gln1291Ter)

NM_000350.2(ABCA4):c.3813G>A (p.Glu1271=)

NM_000350.2(ABCA4):c.1293G>A (p.Trp431Ter)

NM_000350.2(ABCA4):c.206G>A (p.Trp69Ter)

NM_000350.2(ABCA4):c.3322C>T (p.Arg1108Cys)

NM_000350.2(ABCA4):c.1804C>T (p.Arg602Trp)

NM_000350.2(ABCA4):c.1937+1G>A

NM_000350.2(ABCA4):c.2564G>A (p.Trp855Ter)

NM_000350.2(ABCA4):c.4234C>T (p.Gln1412Ter)

NM_000350.2(ABCA4):c.4457C>T (p.Pro1486Leu)

NM_000350.2(ABCA4):c.4594G>A (p.Asp1532Asn)

NM_000350.2(ABCA4):c.4919G>A (p.Arg1640Gln)

NM_000350.2(ABCA4):c.5196+1G>A

NM_000350.2(ABCA4):c.6316C>T (p.Arg2106Cys)

NM_000350.2(ABCA4):c.3056C>T (p.Thr1019Met)

NM_000350.2(ABCA4):c.52C>T (p.Arg18Trp)

NM_000350.2(ABCA4):c.122G>A (p.Trp41Ter)

NM_000350.2(ABCA4):c.1903C>T (p.Gln635Ter)

NM_000350.2(ABCA4):c.194G>A (p.Gly65Glu)

NM_000350.2(ABCA4):c.3085C>T (p.Gln1029Ter)

NM_000350.2(ABCA4):c.4195G>A (p.Glu1399Lys)

NM_000350.2(ABCA4):c.454C>T (p.Arg152Ter)

NM_000350.2(ABCA4):c.45G>A (p.Trp15Ter)

NM_000350.2(ABCA4):c.4610C>T (p.Thr1537Met)

NM_000350.2(ABCA4):c.6112C>T (p.Arg2038Trp)

NM_000350.2(ABCA4):c.6118C>T (p.Arg2040Ter)

NM_000350.2(ABCA4):c.6342G>A (p.Val2114=)

NM_000350.2(ABCA4):c.6658C>T (p.Gln2220Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Stargardt Disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the ABCA4 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00875] Bardet-Biedl Syndrome

[00876] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Bardet-Biedl Syndrome. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from BBS1, BBS2, BBS7, BBS9, BBS10, BBS12, LZTFL1, and TRIM32, including at least the followings:

NM_024649.4(BBS1):c.416G>A (p.Trp139Ter)

NM_024649.4(BBS1):c.871C>T (p.Gln291Ter)

NM_198428.2(BBS9):c.263+1G>A

NM_001178007.1(BBS12):c.1704G>A (p.Trp568Ter)

NM_001276378.1(LZTFL1):c.271C>T (p.Arg91Ter)

NM_031885.3(BBS2):c.1864C>T (p.Arg622Ter)

NM_198428.2(BBS9):c.1759C>T (p.Arg587Ter)

NM_198428.2(BBS9):c.1789+1G>A

NM_024649.4(BBS1):c.432+1G>A

NM_176824.2(BBS7):c.632C>T (p.Thr211Ile)

NM_012210.3(TRIM32):c.388C>T (p.Pro130Ser)

NM_031885.3(BBS2):c.823C>T (p.Arg275Ter)

NM_024685.3(BBS10):c.145C>T (p.Arg49Trp)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Bardet-Biedl Syndrome by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from BBS1, BBS2, BBS7, BBS9, BBS10, BBS12, LZTFL1, and TRIM32, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00877] Cone-rod dystrophy

[00878] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Cone-rod dystrophy. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from RPGRIP1, DRAM2, ABCA4, ADAM9, and CACNA1F, including at least the followings:

NM_020366.3(RPGRIP1):c.154C>T (p.Arg52Ter)

NM_178454.5(DRAM2):c.494G>A (p.Trp165Ter)

NM_178454.5(DRAM2):c.131G>A (p.Ser44Asn)

NM_000350.2(ABCA4):c.161G>A (p.Cys54Tyr)

NM_000350.2(ABCA4):c.5714+5G>A

NM_000350.2(ABCA4):c.880C>T (p.Gln294Ter)

NM_000350.2(ABCA4):c.6079C>T (p.Leu2027Phe)

NM_000350.2(ABCA4):c.3113C>T (p.Ala1038Val)

NM_000350.2(ABCA4):c.634C>T (p.Arg212Cys)

NM_003816.2(ADAM9):c.490C>T (p.Arg164Ter)

NM_005183.3(CACNA1F):c.244C>T (p.Arg82Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Cone-rod dystrophy by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from RPGRIP1, DRAM2, ABCA4, ADAM9, and CACNA1F, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00879] Congenital Stationary Night Blindness

[00880] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Congenital Stationary Night Blindness. In some embodiment, the pathogenic

mutations/SNPs are present in at least one gene selected from GRM6, TRPM1, GPR179, and CACNA1F, including at least the followings:

NM_000843.3(GRM6):c.1462C>T (p.Gln488Ter)

NM_002420.5(TRPM1):c.2998C>T (p.Arg1000Ter)

NM_001004334.3(GPR179):c.673C>T (p.Gln225Ter)

NM_005183.3(CACNA1F):c.2576+1G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Congenital Stationary Night Blindness by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from GRM6, TRPM1, GPR179, and CACNA1F, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00881] Usher Syndrome

[00882] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Usher Syndrome. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from MYO7A, USH1C, CDH23, PCDH15, USH2A, ADGRV1, WHRN, and CLRN1, including at least the followings:

NM_000260.3(MYO7A):c.640G>A (p.Gly214Arg)

NM_000260.3(MYO7A):c.1200+1G>A

NM_000260.3(MYO7A):c.141G>A (p.Trp47Ter)

NM_000260.3(MYO7A):c.1556G>A (p.Gly519Asp)

NM_000260.3(MYO7A):c.1900C>T (p.Arg634Ter)

NM_000260.3(MYO7A):c.1963C>T (p.Gln655Ter)

NM_000260.3(MYO7A):c.2094+1G>A

NM_000260.3(MYO7A):c.4293G>A (p.Trp1431Ter)

NM_000260.3(MYO7A):c.5101C>T (p.Arg1701Ter)

NM_000260.3(MYO7A):c.5617C>T (p.Arg1873Trp)

NM_000260.3(MYO7A):c.5660C>T (p.Pro1887Leu)

NM_000260.3(MYO7A):c.6070C>T (p.Arg2024Ter)

NM_000260.3(MYO7A):c.470+1G>A

NM_000260.3(MYO7A):c.5968C>T (p.Gln1990Ter)

NM_000260.3(MYO7A):c.3719G>A (p.Arg1240Gln)

NM_000260.3(MYO7A):c.494C>T (p.Thr165Met)

NM_000260.3(MYO7A):c.5392C>T (p.Gln1798Ter)

NM_000260.3(MYO7A):c.5648G>A (p.Arg1883Gln)

NM_000260.3(MYO7A):c.448C>T (p.Arg150Ter)

NM_000260.3(MYO7A):c.700C>T (p.Gln234Ter)

NM_000260.3(MYO7A):c.635G>A (p.Arg212His)

NM_000260.3(MYO7A):c.1996C>T (p.Arg666Ter)

NM_005709.3(USH1C):c.216G>A (p.Val72=)

NM_022124.5(CDH23):c.7362+5G>A

NM_022124.5(CDH23):c.3481C>T (p.Arg1161Ter)

NM_022124.5(CDH23):c.3628C>T (p.Gln1210Ter)

NM_022124.5(CDH23):c.5272C>T (p.Gln1758Ter)

NM_022124.5(CDH23):c.5712+1G>A

NM_022124.5(CDH23):c.5712G>A (p.Thr1904=)

NM_022124.5(CDH23):c.5923+1G>A

NM_022124.5(CDH23):c.6049+1G>A

NM_022124.5(CDH23):c.7776G>A (p.Trp2592Ter)

NM_022124.5(CDH23):c.9556C>T (p.Arg3186Ter)

NM_022124.5(CDH23):c.3706C>T (p.Arg1236Ter)

NM_022124.5(CDH23):c.4309C>T (p.Arg1437Ter)

NM_022124.5(CDH23):c.6050-9G>A

NM_033056.3(PCDH15):c.3316C>T (p.Arg1106Ter)

NM_033056.3(PCDH15):c.7C>T (p.Arg3Ter)

NM_033056.3(PCDH15):c.1927C>T (p.Arg643Ter)

NM_001142772.1(PCDH15):c.400C>T (p.Arg134Ter)

NM_033056.3(PCDH15):c.3358C>T (p.Arg1120Ter)

NM_206933.2(USH2A):c.11048-1G>A

NM_206933.2(USH2A):c.1143+1G>A

NM_206933.2(USH2A):c.11954G>A (p.Trp3985Ter)

NM_206933.2(USH2A):c.12868C>T (p.Gln4290Ter)

NM_206933.2(USH2A):c.14180G>A (p.Trp4727Ter)

NM_206933.2(USH2A):c.14911C>T (p.Arg4971Ter)

NM_206933.2(USH2A):c.5788C>T (p.Arg1930Ter)

NM_206933.2(USH2A):c.5858-1G>A

NM_206933.2(USH2A):c.6224G>A (p.Trp2075Ter)

NM_206933.2(USH2A):c.820C>T (p.Arg274Ter)
NM_206933.2(USH2A):c.8981G>A (p.Trp2994Ter)
NM_206933.2(USH2A):c.9304C>T (p.Gln3102Ter)
NM_206933.2(USH2A):c.13010C>T (p.Thr4337Met)
NM_206933.2(USH2A):c.14248C>T (p.Gln4750Ter)
NM_206933.2(USH2A):c.6398G>A (p.Trp2133Ter)
NM_206933.2(USH2A):c.632G>A (p.Trp211Ter)
NM_206933.2(USH2A):c.6601C>T (p.Gln2201Ter)
NM_206933.2(USH2A):c.13316C>T (p.Thr4439Ile)
NM_206933.2(USH2A):c.4405C>T (p.Gln1469Ter)
NM_206933.2(USH2A):c.9570+1G>A
NM_206933.2(USH2A):c.8740C>T (p.Arg2914Ter)
NM_206933.2(USH2A):c.8681+1G>A
NM_206933.2(USH2A):c.1000C>T (p.Arg334Trp)
NM_206933.2(USH2A):c.14175G>A (p.Trp4725Ter)
NM_206933.2(USH2A):c.9390G>A (p.Trp3130Ter)
NM_206933.2(USH2A):c.908G>A (p.Arg303His)
NM_206933.2(USH2A):c.5776+1G>A
NM_206933.2(USH2A):c.11156G>A (p.Arg3719His)
NM_032119.3(ADGRV1):c.2398C>T (p.Arg800Ter)
NM_032119.3(ADGRV1):c.7406G>A (p.Trp2469Ter)
NM_032119.3(ADGRV1):c.12631C>T (p.Arg4211Ter)
NM_032119.3(ADGRV1):c.7129C>T (p.Arg2377Ter)
NM_032119.3(ADGRV1):c.14885G>A (p.Trp4962Ter)
NM_015404.3(WHRN):c.1267C>T (p.Arg423Ter)
NM_174878.2(CLRN1):c.619C>T (p.Arg207Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Enhanced Usher Syndrome by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from MYO7A, USH1C, CDH23, PCDH15, USH2A, ADGRV1, WHRN, and CLRN1, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00883] Leber Congenital Amaurosis

[00884] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Leber Congenital Amaurosis. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from TULP1, RPE65, SPATA7, AIPL1, CRB1, NMNAT1, and PEX1, including at least the followings:

NM_003322.5(TULP1):c.1495+1G>A

NM_000329.2(RPE65):c.11+5G>A

NM_018418.4(SPATA7):c.322C>T (p.Arg108Ter)

NM_014336.4(AIPL1):c.784G>A (p.Gly262Ser)

NM_201253.2(CRB1):c.1576C>T (p.Arg526Ter)

NM_201253.2(CRB1):c.3307G>A (p.Gly1103Arg)

NM_201253.2(CRB1):c.2843G>A (p.Cys948Tyr)

NM_022787.3(NMNAT1):c.769G>A (p.Glu257Lys)

NM_000466.2(PEX1):c.2528G>A (p.Gly843Asp)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Leber Congenital Amaurosis by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from TULP1, RPE65, SPATA7, AIPL1, CRB1, NMNAT1, and PEX1, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00885] Retinitis Pigmentosa

[00886] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Retinitis Pigmentosa. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from CRB1, IFT140, RP1, IMPDH1, PRPF31, RPGR, ABCA4, RPE65, EYS, NRL, FAM161A, NR2E3, USH2A, RHO, PDE6B, KLHL7, PDE6A, CNGB1, BEST1, C2orf71, PRPH2, CA4, CERKL, RPE65, PDE6B, and ADGRV1, including at least the followings:

NM_001257965.1(CRB1):c.2711G>A (p.Cys904Tyr)

NM_014714.3(IFT140):c.3827G>A (p.Gly1276Glu)

NM_006269.1(RP1):c.2029C>T (p.Arg677Ter)

NM_000883.3(IMPDH1):c.931G>A (p.Asp311Asn)

NM_015629.3(PRPF31):c.1273C>T (p.Gln425Ter)

NM_015629.3(PRPF31):c.1073+1G>A

NM_000328.2(RPGR):c.1387C>T (p.Gln463Ter)

NM_000350.2(ABCA4):c.4577C>T (p.Thr1526Met)

NM_000350.2(ABCA4):c.6229C>T (p.Arg2077Trp)

NM_000329.2(RPE65):c.271C>T (p.Arg91Trp)

NM_001142800.1(EYS):c.2194C>T (p.Gln732Ter)

NM_001142800.1(EYS):c.490C>T (p.Arg164Ter)

NM_006177.3(NRL):c.151C>T (p.Pro51Ser)

NM_001201543.1(FAM161A):c.1567C>T (p.Arg523Ter)

NM_014249.3(NR2E3):c.166G>A (p.Gly56Arg)

NM_206933.2(USH2A):c.2209C>T (p.Arg737Ter)

NM_206933.2(USH2A):c.14803C>T (p.Arg4935Ter)

NM_206933.2(USH2A):c.10073G>A (p.Cys3358Tyr)

NM_000539.3(RHO):c.541G>A (p.Glu181Lys)

NM_000283.3(PDE6B):c.892C>T (p.Gln298Ter)

NM_001031710.2(KLHL7):c.458C>T (p.Ala153Val)

NM_000440.2(PDE6A):c.1926+1G>A

NM_001297.4(CNGB1):c.2128C>T (p.Gln710Ter)

NM_001297.4(CNGB1):c.952C>T (p.Gln318Ter)

NM_004183.3(BEST1):c.682G>A (p.Asp228Asn)

NM_001029883.2(C2orf71):c.1828C>T (p.Gln610Ter)

NM_000322.4(PRPH2):c.647C>T (p.Pro216Leu)

NM_000717.4(CA4):c.40C>T (p.Arg14Trp)

NM_201548.4(CERKL):c.769C>T (p.Arg257Ter)

NM_000329.2(RPE65):c.118G>A (p.Gly40Ser)

NM_000322.4(PRPH2):c.499G>A (p.Gly167Ser)

NM_000539.3(RHO):c.403C>T (p.Arg135Trp)

NM_000283.3(PDE6B):c.2193+1G>A

NM_032119.3(ADGRV1):c.6901C>T (p.Gln2301Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Retinitis Pigmentosa by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from CRB1, IFT140, RP1, IMPDH1, PRPF31, RPGR, ABCA4, RPE65, EYS, NRL, FAM161A, NR2E3, USH2A, RHO, PDE6B, KLHL7, PDE6A, CNGB1, BEST1, C2orf71, PRPH2, CA4, CERKL, RPE65, PDE6B, and

ADGRV1, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00887] Achromatopsia

[00888] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Achromatopsia. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from CNGA3, CNGB3, and ATF6, including at least the followings:
NM_001298.2(CNGA3):c.847C>T (p.Arg283Trp)
NM_001298.2(CNGA3):c.101+1G>A
NM_001298.2(CNGA3):c.1585G>A (p.Val529Met)
NM_019098.4(CNGB3):c.1578+1G>A
NM_019098.4(CNGB3):c.607C>T (p.Arg203Ter)
NM_019098.4(CNGB3):c.1119G>A (p.Trp373Ter)
NM_007348.3(ATF6):c.970C>T (p.Arg324Cys)
See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Achromatopsia by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from CNGA3, CNGB3, and ATF6, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00889] **Diseases Affecting Hearing**

[00890] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various diseases affecting hearing are reported in the ClinVar database and disclosed in Table A, including but not limited to deafness and Nonsyndromic hearing loss. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00891] Deafness

[00892] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with deafness. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from FGF3, MYO7A, STRC, ACTG1, SLC17A8, TMC1, GJB2, MYH14, COCH, CDH23, USH1C, GJB2, MYO7A, PCDH15, MYO15A, MYO3A, WHRN, DFNB59, TMC1, LOXHD1, TMPRSS3, OTOGL, OTOF, JAG1, and MARVELD2, including at least the followings:
NM_005247.2(FGF3):c.283C>T (p.Arg95Trp)

NM_000260.3(MYO7A):c.652G>A (p.Asp218Asn)

NM_000260.3(MYO7A):c.689C>T (p.Ala230Val)

NM_153700.2(STRC):c.4057C>T (p.Gln1353Ter)

NM_001614.3(ACTG1):c.721G>A (p.Glu241Lys)

NM_139319.2(SLC17A8):c.632C>T (p.Ala211Val)

NM_138691.2(TMC1):c.1714G>A (p.Asp572Asn)

NM_004004.5(GJB2):c.598G>A (p.Gly200Arg)

NM_004004.5(GJB2):c.71G>A (p.Trp24Ter)

NM_004004.5(GJB2):c.416G>A (p.Ser139Asn)

NM_004004.5(GJB2):c.224G>A (p.Arg75Gln)

NM_004004.5(GJB2):c.95G>A (p.Arg32His)

NM_004004.5(GJB2):c.250G>A (p.Val84Met)

NM_004004.5(GJB2):c.428G>A (p.Arg143Gln)

NM_004004.5(GJB2):c.551G>A (p.Arg184Gln)

NM_004004.5(GJB2):c.223C>T (p.Arg75Trp)

NM_024729.3(MYH14):c.359C>T (p.Ser120Leu)

NM_004086.2(COCH):c.151C>T (p.Pro51Ser)

NM_022124.5(CDH23):c.4021G>A (p.Asp1341Asn)

NM_153700.2(STRC):c.4701+1G>A

NM_153676.3(USH1C):c.496+1G>A

NM_004004.5(GJB2):c.131G>A (p.Trp44Ter)

NM_004004.5(GJB2):c.283G>A (p.Val95Met)

NM_004004.5(GJB2):c.298C>T (p.His100Tyr)

NM_004004.5(GJB2):c.427C>T (p.Arg143Trp)

NM_004004.5(GJB2):c.109G>A (p.Val37Ile)

NM_004004.5(GJB2):c.-23+1G>A

NM_004004.5(GJB2):c.148G>A (p.Asp50Asn)

NM_004004.5(GJB2):c.134G>A (p.Gly45Glu)

NM_004004.5(GJB2):c.370C>T (p.Gln124Ter)

NM_004004.5(GJB2):c.230G>A (p.Trp77Ter)

NM_004004.5(GJB2):c.231G>A (p.Trp77Ter)

NM_000260.3(MYO7A):c.5899C>T (p.Arg1967Ter)

NM_000260.3(MYO7A):c.2005C>T (p.Arg669Ter)

NM_033056.3(PCDH15):c.733C>T (p.Arg245Ter)

NM_016239.3(MYO15A):c.3866+1G>A

NM_016239.3(MYO15A):c.6178-1G>A

NM_016239.3(MYO15A):c.8714-1G>A

NM_017433.4(MYO3A):c.2506-1G>A

NM_015404.3(WHRN):c.1417-1G>A

NM_001042702.3(DFNB59):c.499C>T (p.Arg167Ter)

NM_138691.2(TMC1):c.100C>T (p.Arg34Ter)

NM_138691.2(TMC1):c.1165C>T (p.Arg389Ter)

NM_144612.6(LOXHD1):c.2008C>T (p.Arg670Ter)

NM_144612.6(LOXHD1):c.4714C>T (p.Arg1572Ter)

NM_144612.6(LOXHD1):c.4480C>T (p.Arg1494Ter)

NM_024022.2(TMPRSS3):c.325C>T (p.Arg109Trp)

NM_173591.3(OTOGL):c.3076C>T (p.Gln1026Ter)

NM_194248.2(OTOF):c.4483C>T (p.Arg1495Ter)

NM_194248.2(OTOF):c.2122C>T (p.Arg708Ter)

NM_194248.2(OTOF):c.2485C>T (p.Gln829Ter)

NM_001038603.2(MARVELD2):c.1498C>T (p.Arg500Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing deafness by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from FGF3, MYO7A, STRC, ACTG1, SLC17A8, TMC1, GJB2, MYH14, COCH, CDH23, USH1C, GJB2, MYO7A, PCDH15, MYO15A, MYO3A, WHRN, DFNB59, TMC1, LOXHD1, TMPRSS3, OTOGL, OTOF, JAG1, and MARVELD2, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00893] Nonsyndromic hearing loss

[00894] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Nonsyndromic hearing loss. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from GJB2, POU3F4, MYO15A, TMPRSS3, LOXHD1, OTOF, MYO6, OTOA, STRC, TRIOBP, MARVELD2, TMC1, TECTA, OTOGL, and GIPC3, including at least the followings:

NM_004004.5(GJB2):c.169C>T (p.Gln57Ter)

NM_000307.4(POU3F4):c.499C>T (p.Arg167Ter)

NM_016239.3(MYO15A):c.8767C>T (p.Arg2923Ter)

NM_024022.2(TMPRSS3):c.323-6G>A

NM_024022.2(TMPRSS3):c.916G>A (p.Ala306Thr)

NM_144612.6(LOXHD1):c.2497C>T (p.Arg833Ter)

NM_194248.2(OTOF):c.2153G>A (p.Trp718Ter)

NM_194248.2(OTOF):c.2818C>T (p.Gln940Ter)

NM_194248.2(OTOF):c.4799+1G>A

NM_004999.3(MYO6):c.826C>T (p.Arg276Ter)

NM_144672.3(OTOA):c.1880+1G>A

NM_153700.2(STRC):c.5188C>T (p.Arg1730Ter)

NM_153700.2(STRC):c.3670C>T (p.Arg1224Ter)

NM_153700.2(STRC):c.4402C>T (p.Arg1468Ter)

NM_024022.2(TMPRSS3):c.1192C>T (p.Gln398Ter)

NM_001039141.2(TRIOBP):c.6598C>T (p.Arg2200Ter)

NM_016239.3(MYO15A):c.7893+1G>A

NM_016239.3(MYO15A):c.5531+1G>A

NM_016239.3(MYO15A):c.6046+1G>A

NM_144612.6(LOXHD1):c.3169C>T (p.Arg1057Ter)

NM_001038603.2(MARVELD2):c.1331+1G>A

NM_138691.2(TMC1):c.1676G>A (p.Trp559Ter)

NM_138691.2(TMC1):c.1677G>A (p.Trp559Ter)

NM_005422.2(TECTA):c.5977C>T (p.Arg1993Ter)

NM_173591.3(OTOGL):c.4987C>T (p.Arg1663Ter)

NM_153700.2(STRC):c.3493C>T (p.Gln1165Ter)

NM_153700.2(STRC):c.3217C>T (p.Arg1073Ter)

NM_016239.3(MYO15A):c.5896C>T (p.Arg1966Ter)

NM_133261.2(GIPC3):c.411+1G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Nonsyndromic hearing loss by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from GJB2, POU3F4, MYO15A, TMPRSS3, LOXHD1, OTOF, MYO6, OTOA, STRC, TRIOBP, MARVELD2, TMC1, TECTA, OTOGL, and GIPC3, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00895] Blood Disorders

[00896] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various blood disorders are reported in the ClinVar database and disclosed in Table A, including but not limited to Beta-thalassemia, Hemophilia A, Hemophilia B, Hemophilia C, and Wiskott-Aldrich syndrome. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00897] Beta-thalassemia

[00898] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Beta-thalassemia. In some embodiment, the pathogenic mutations/SNPs are present in at least the HBB gene, including at least the followings:

NM_000518.4(HBB):c.-137C>T

NM_000518.4(HBB):c.-50-88C>T

NM_000518.4(HBB):c.-140C>T

NM_000518.4(HBB):c.316-197C>T

NM_000518.4(HBB):c.93-21G>A

NM_000518.4(HBB):c.114G>A (p.Trp38Ter)

NM_000518.4(HBB):c.118C>T (p.Gln40Ter)

NM_000518.4(HBB):c.92+1G>A

NM_000518.4(HBB):c.315+1G>A

NM_000518.4(HBB):c.92+5G>A

NM_000518.4(HBB):c.-50-101C>T

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Beta-thalassemia by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the HBB gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00899] Hemophilia A

[00900] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Hemophilia A. In some embodiment, the pathogenic mutations/SNPs are present in at least the F8 gene, including at least the followings:

NM_000132.3(F8):c.3169G>A (p.Glu1057Lys)

NM_000132.3(F8):c.902G>A (p.Arg301His)

NM_000132.3(F8):c.1834C>T (p.Arg612Cys)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Hemophilia A by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the F8 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00901] Hemophilia B

[00902] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Hemophilia B. In some embodiment, the pathogenic mutations/SNPs are present in at least the F9 gene, including at least the followings:

NM_000133.3(F9):c.835G>A (p.Ala279Thr)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Hemophilia B by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the F9 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00903] Hemophilia C

[00904] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Hemophilia C. In some embodiment, the pathogenic mutations/SNPs are present in at least the F11 gene, including at least the followings:

NM_000128.3(F11):c.400C>T (p.Gln134Ter)

NM_000128.3(F11):c.1432G>A (p.Gly478Arg)

NM_000128.3(F11):c.1288G>A (p.Ala430Thr)

NM_000128.3(F11):c.326-1G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Hemophilia C by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the F11 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00905] Wiskott-Aldrich syndrome

[00906] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Wiskott-Aldrich syndrome. In some embodiment, the pathogenic mutations/SNPs are present in at least the WAS gene, including at least the followings:

NM_000377.2(WAS):c.37C>T (p.Arg13Ter)

NM_000377.2(WAS):c.257G>A (p.Arg86His)

NM_000377.2(WAS):c.777+1G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Wiskott-Aldrich syndrome by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the WAS gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00907] **Liver Diseases**

[00908] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various liver diseases are reported in the ClinVar database and disclosed in Table A, including but not limited to Transthyretin amyloidosis, Alpha-1-antitrypsin deficiency, Wilson's disease, and Phenylketonuria. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00909] Transthyretin amyloidosis

[00910] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Transthyretin amyloidosis. In some embodiment, the pathogenic mutations/SNPs are present in at least the TTR gene, including at least the followings:

NM_000371.3(TTR):c.424G>A (p.Val142Ile)

NM_000371.3(TTR):c.148G>A (p.Val50Met)

NM_000371.3(TTR):c.118G>A (p.Val40Ile)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Transthyretin amyloidosis by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the TTR gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00911] Alpha-1-antitrypsin deficiency

[00912] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Alpha-1-antitrypsin deficiency. In some embodiment, the pathogenic mutations/SNPs are present in at least the SERPINA1 gene, including at least the followings:

NM_000295.4(SERPINA1):c.538C>T (p.Gln180Ter)

NM_001127701.1(SERPINA1):c.1178C>T (p.Pro393Leu)

NM_001127701.1(SERPINA1):c.230C>T (p.Ser77Phe)

NM_001127701.1(SERPINA1):c.1096G>A (p.Glu366Lys)

NM_000295.4(SERPINA1):c.1177C>T (p.Pro393Ser)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Alpha-1-antitrypsin deficiency by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the SERPINA1 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00913] Wilson's disease

[00914] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Wilson's disease. In some embodiment, the pathogenic mutations/SNPs are present in at least the ATP7B gene, including at least the followings:

NM_000053.3(ATP7B):c.2293G>A (p.Asp765Asn)

NM_000053.3(ATP7B):c.3955C>T (p.Arg1319Ter)

NM_000053.3(ATP7B):c.2865+1G>A

NM_000053.3(ATP7B):c.3796G>A (p.Gly1266Arg)

NM_000053.3(ATP7B):c.2621C>T (p.Ala874Val)

NM_000053.3(ATP7B):c.2071G>A (p.Gly691Arg)

NM_000053.3(ATP7B):c.2128G>A (p.Gly710Ser)

NM_000053.3(ATP7B):c.2336G>A (p.Trp779Ter)

NM_000053.3(ATP7B):c.4021G>A (p.Gly1341Ser)

NM_000053.3(ATP7B):c.3182G>A (p.Gly1061Glu)

NM_000053.3(ATP7B):c.4114C>T (p.Gln1372Ter)

NM_000053.3(ATP7B):c.1708-1G>A

NM_000053.3(ATP7B):c.865C>T (p.Gln289Ter)

NM_000053.3(ATP7B):c.2930C>T (p.Thr977Met)

NM_000053.3(ATP7B):c.3659C>T (p.Thr1220Met)

NM_000053.3(ATP7B):c.2605G>A (p.Gly869Arg)

NM_000053.3(ATP7B):c.2975C>T (p.Pro992Leu)

NM_000053.3(ATP7B):c.2519C>T (p.Pro840Leu)

NM_000053.3(ATP7B):c.2906G>A (p.Arg969Gln)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Wilson's disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the ATP7B gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00915] Phenylketonuria

[00916] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Phenylketonuria. In some embodiment, the pathogenic mutations/SNPs are present in at least the PAH gene, including at least the followings:

NM_000277.1(PAH):c.1315+1G>A

NM_000277.1(PAH):c.1222C>T (p.Arg408Trp)

NM_000277.1(PAH):c.838G>A (p.Glu280Lys)

NM_000277.1(PAH):c.331C>T (p.Arg111Ter)

NM_000277.1(PAH):c.782G>A (p.Arg261Gln)

NM_000277.1(PAH):c.754C>T (p.Arg252Trp)

NM_000277.1(PAH):c.473G>A (p.Arg158Gln)

NM_000277.1(PAH):c.727C>T (p.Arg243Ter)

NM_000277.1(PAH):c.842C>T (p.Pro281Leu)

NM_000277.1(PAH):c.728G>A (p.Arg243Gln)

NM_000277.1(PAH):c.1066-11G>A

NM_000277.1(PAH):c.781C>T (p.Arg261Ter)

NM_000277.1(PAH):c.1223G>A (p.Arg408Gln)

NM_000277.1(PAH):c.1162G>A (p.Val388Met)

NM_000277.1(PAH):c.1066-3C>T

NM_000277.1(PAH):c.1208C>T (p.Ala403Val)

NM_000277.1(PAH):c.890G>A (p.Arg297His)

NM_000277.1(PAH):c.926C>T (p.Ala309Val)

NM_000277.1(PAH):c.441+1G>A

NM_000277.1(PAH):c.526C>T (p.Arg176Ter)

NM_000277.1(PAH):c.688G>A (p.Val230Ile)

NM_000277.1(PAH):c.721C>T (p.Arg241Cys)

NM_000277.1(PAH):c.745C>T (p.Leu249Phe)

NM_000277.1(PAH):c.442-1G>A

NM_000277.1(PAH):c.842+1G>A

NM_000277.1(PAH):c.776C>T (p.Ala259Val)

NM_000277.1(PAH):c.1200-1G>A

NM_000277.1(PAH):c.912+1G>A

NM_000277.1(PAH):c.1065+1G>A

NM_000277.1(PAH):c.472C>T (p.Arg158Trp)

NM_000277.1(PAH):c.755G>A (p.Arg252Gln)

NM_000277.1(PAH):c.809G>A (p.Arg270Lys)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Phenylketonuria by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the PAH gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00917] Kidney Diseases

[00918] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various kidney diseases are reported in the ClinVar database and disclosed in Table A, including but not limited to Autosomal recessive polycystic kidney disease and Renal carnitine transport defect. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00919] Autosomal recessive polycystic kidney disease

[00920] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Autosomal recessive polycystic kidney disease. In some embodiment, the pathogenic mutations/SNPs are present in at least the PKHD1 gene, including at least the followings:

NM_138694.3(PKHD1):c.10444C>T (p.Arg3482Cys)

NM_138694.3(PKHD1):c.9319C>T (p.Arg3107Ter)

NM_138694.3(PKHD1):c.1480C>T (p.Arg494Ter)

NM_138694.3(PKHD1):c.707+1G>A

NM_138694.3(PKHD1):c.1486C>T (p.Arg496Ter)

NM_138694.3(PKHD1):c.8303-1G>A

NM_138694.3(PKHD1):c.2854G>A (p.Gly952Arg)

NM_138694.3(PKHD1):c.7194G>A (p.Trp2398Ter)

NM_138694.3(PKHD1):c.10219C>T (p.Gln3407Ter)

NM_138694.3(PKHD1):c.107C>T (p.Thr36Met)

NM_138694.3(PKHD1):c.8824C>T (p.Arg2942Ter)

NM_138694.3(PKHD1):c.982C>T (p.Arg328Ter)

NM_138694.3(PKHD1):c.4870C>T (p.Arg1624Trp)

NM_138694.3(PKHD1):c.1602+1G>A

NM_138694.3(PKHD1):c.1694-1G>A

NM_138694.3(PKHD1):c.2341C>T (p.Arg781Ter)

NM_138694.3(PKHD1):c.2407+1G>A

NM_138694.3(PKHD1):c.2452C>T (p.Gln818Ter)

NM_138694.3(PKHD1):c.5236+1G>A

NM_138694.3(PKHD1):c.6499C>T (p.Gln2167Ter)

NM_138694.3(PKHD1):c.2725C>T (p.Arg909Ter)

NM_138694.3(PKHD1):c.370C>T (p.Arg124Ter)

NM_138694.3(PKHD1):c.2810G>A (p.Trp937Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Autosomal recessive polycystic kidney disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the PKHD1 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00921] Renal carnitine transport defect

[00922] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Renal carnitine transport defect. In some embodiment, the pathogenic mutations/SNPs are present in at least the SLC22A5 gene, including at least the followings:

NM_003060.3(SLC22A5):c.760C>T (p.Arg254Ter)

NM_003060.3(SLC22A5):c.396G>A (p.Trp132Ter)

NM_003060.3(SLC22A5):c.844C>T (p.Arg282Ter)

NM_003060.3(SLC22A5):c.505C>T (p.Arg169Trp)

NM_003060.3(SLC22A5):c.1319C>T (p.Thr440Met)

NM_003060.3(SLC22A5):c.1195C>T (p.Arg399Trp)

NM_003060.3(SLC22A5):c.695C>T (p.Thr232Met)

NM_003060.3(SLC22A5):c.845G>A (p.Arg282Gln)

NM_003060.3(SLC22A5):c.1193C>T (p.Pro398Leu)

NM_003060.3(SLC22A5):c.1463G>A (p.Arg488His)

NM_003060.3(SLC22A5):c.338G>A (p.Cys113Tyr)

NM_003060.3(SLC22A5):c.136C>T (p.Pro46Ser)

NM_003060.3(SLC22A5):c.506G>A (p.Arg169Gln)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Renal carnitine transport defect by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the SLC22A5 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00923] Muscle Diseases

[00924] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various muscle diseases are reported in the ClinVar database and disclosed in Table A, including but not limited to Duchenne muscular dystrophy, Becker muscular dystrophy, Limb-girdle muscular dystrophy, Emery-Dreifuss muscular dystrophy, and Facioscapulohumeral muscular dystrophy. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00925] Duchenne muscular dystrophy

[00926] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Duchenne muscular dystrophy. In some embodiment, the pathogenic mutations/SNPs are present in at least the DMD gene, including at least the followings:

NM_004006.2(DMD):c.2797C>T (p.Gln933Ter)

NM_004006.2(DMD):c.4870C>T (p.Gln1624Ter)

NM_004006.2(DMD):c.5551C>T (p.Gln1851Ter)

NM_004006.2(DMD):c.3188G>A (p.Trp1063Ter)

NM_004006.2(DMD):c.8357G>A (p.Trp2786Ter)

NM_004006.2(DMD):c.7817G>A (p.Trp2606Ter)

NM_004006.2(DMD):c.7755G>A (p.Trp2585Ter)

NM_004006.2(DMD):c.5917C>T (p.Gln1973Ter)

NM_004006.2(DMD):c.5641C>T (p.Gln1881Ter)

NM_004006.2(DMD):c.5131C>T (p.Gln1711Ter)
NM_004006.2(DMD):c.4240C>T (p.Gln1414Ter)
NM_004006.2(DMD):c.3427C>T (p.Gln1143Ter)
NM_004006.2(DMD):c.2407C>T (p.Gln803Ter)
NM_004006.2(DMD):c.2368C>T (p.Gln790Ter)
NM_004006.2(DMD):c.1683G>A (p.Trp561Ter)
NM_004006.2(DMD):c.1663C>T (p.Gln555Ter)
NM_004006.2(DMD):c.1388G>A (p.Trp463Ter)
NM_004006.2(DMD):c.1331+1G>A
NM_004006.2(DMD):c.1324C>T (p.Gln442Ter)
NM_004006.2(DMD):c.355C>T (p.Gln119Ter)
NM_004006.2(DMD):c.94-1G>A
NM_004006.2(DMD):c.5506C>T (p.Gln1836Ter)
NM_004006.2(DMD):c.1504C>T (p.Gln502Ter)
NM_004006.2(DMD):c.5032C>T (p.Gln1678Ter)
NM_004006.2(DMD):c.457C>T (p.Gln153Ter)
NM_004006.2(DMD):c.1594C>T (p.Gln532Ter)
NM_004006.2(DMD):c.1150-1G>A
NM_004006.2(DMD):c.6223C>T (p.Gln2075Ter)
NM_004006.2(DMD):c.3747G>A (p.Trp1249Ter)
NM_004006.2(DMD):c.2861G>A (p.Trp954Ter)
NM_004006.2(DMD):c.9563+1G>A
NM_004006.2(DMD):c.4483C>T (p.Gln1495Ter)
NM_004006.2(DMD):c.4312C>T (p.Gln1438Ter)
NM_004006.2(DMD):c.8209C>T (p.Gln2737Ter)
NM_004006.2(DMD):c.4071+1G>A
NM_004006.2(DMD):c.2665C>T (p.Arg889Ter)
NM_004006.2(DMD):c.2202G>A (p.Trp734Ter)
NM_004006.2(DMD):c.2077C>T (p.Gln693Ter)
NM_004006.2(DMD):c.1653G>A (p.Trp551Ter)
NM_004006.2(DMD):c.1061G>A (p.Trp354Ter)
NM_004006.2(DMD):c.8914C>T (p.Gln2972Ter)
NM_004006.2(DMD):c.6118-1G>A
NM_004006.2(DMD):c.4729C>T (p.Arg1577Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Duchenne muscular dystrophy by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the DMD gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00927] Becker muscular dystrophy

[00928] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Becker muscular dystrophy. In some embodiment, the pathogenic mutations/SNPs are present in at least the DMD gene, including at least the followings:

NM_004006.2(DMD):c.3413G>A (p.Trp1138Ter)

NM_004006.2(DMD):c.358-1G>A

NM_004006.2(DMD):c.10108C>T (p.Arg3370Ter)

NM_004006.2(DMD):c.6373C>T (p.Gln2125Ter)

NM_004006.2(DMD):c.9568C>T (p.Arg3190Ter)

NM_004006.2(DMD):c.8713C>T (p.Arg2905Ter)

NM_004006.2(DMD):c.1615C>T (p.Arg539Ter)

NM_004006.2(DMD):c.3151C>T (p.Arg1051Ter)

NM_004006.2(DMD):c.3432+1G>A

NM_004006.2(DMD):c.5287C>T (p.Arg1763Ter)

NM_004006.2(DMD):c.5530C>T (p.Arg1844Ter)

NM_004006.2(DMD):c.8608C>T (p.Arg2870Ter)

NM_004006.2(DMD):c.8656C>T (p.Gln2886Ter)

NM_004006.2(DMD):c.8944C>T (p.Arg2982Ter)

NM_004006.2(DMD):c.5899C>T (p.Arg1967Ter)

NM_004006.2(DMD):c.10033C>T (p.Arg3345Ter)

NM_004006.2(DMD):c.10086+1G>A

NM_004019.2(DMD):c.1020G>A (p.Thr340=)

NM_004006.2(DMD):c.1261C>T (p.Gln421Ter)

NM_004006.2(DMD):c.1465C>T (p.Gln489Ter)

NM_004006.2(DMD):c.1990C>T (p.Gln664Ter)

NM_004006.2(DMD):c.2032C>T (p.Gln678Ter)

NM_004006.2(DMD):c.2332C>T (p.Gln778Ter)

NM_004006.2(DMD):c.2419C>T (p.Gln807Ter)

NM_004006.2(DMD):c.2650C>T (p.Gln884Ter)

NM_004006.2(DMD):c.2804-1G>A

NM_004006.2(DMD):c.3276+1G>A

NM_004006.2(DMD):c.3295C>T (p.Gln1099Ter)

NM_004006.2(DMD):c.336G>A (p.Trp112Ter)

NM_004006.2(DMD):c.3580C>T (p.Gln1194Ter)

NM_004006.2(DMD):c.4117C>T (p.Gln1373Ter)

NM_004006.2(DMD):c.649+1G>A

NM_004006.2(DMD):c.6906G>A (p.Trp2302Ter)

NM_004006.2(DMD):c.7189C>T (p.Gln2397Ter)

NM_004006.2(DMD):c.7309+1G>A

NM_004006.2(DMD):c.7657C>T (p.Arg2553Ter)

NM_004006.2(DMD):c.7682G>A (p.Trp2561Ter)

NM_004006.2(DMD):c.7683G>A (p.Trp2561Ter)

NM_004006.2(DMD):c.7894C>T (p.Gln2632Ter)

NM_004006.2(DMD):c.9361+1G>A

NM_004006.2(DMD):c.9564-1G>A

NM_004006.2(DMD):c.2956C>T (p.Gln986Ter)

NM_004006.2(DMD):c.883C>T (p.Arg295Ter)

NM_004006.2(DMD):c.31+36947G>A

NM_004006.2(DMD):c.10279C>T (p.Gln3427Ter)

NM_004006.2(DMD):c.433C>T (p.Arg145Ter)

NM_004006.2(DMD):c.9G>A (p.Trp3Ter)

NM_004006.2(DMD):c.10171C>T (p.Arg3391Ter)

NM_004006.2(DMD):c.583C>T (p.Arg195Ter)

NM_004006.2(DMD):c.9337C>T (p.Arg3113Ter)

NM_004006.2(DMD):c.8038C>T (p.Arg2680Ter)

NM_004006.2(DMD):c.1812+1G>A

NM_004006.2(DMD):c.1093C>T (p.Gln365Ter)

NM_004006.2(DMD):c.1704+1G>A

NM_004006.2(DMD):c.1912C>T (p.Gln638Ter)

NM_004006.2(DMD):c.133C>T (p.Gln45Ter)

NM_004006.2(DMD):c.5868G>A (p.Trp1956Ter)

NM_004006.2(DMD):c.565C>T (p.Gln189Ter)

NM_004006.2(DMD):c.5089C>T (p.Gln1697Ter)

NM_004006.2(DMD):c.2512C>T (p.Gln838Ter)

NM_004006.2(DMD):c.10477C>T (p.Gln3493Ter)

NM_004006.2(DMD):c.93+1G>A

NM_004006.2(DMD):c.4174C>T (p.Gln1392Ter)

NM_004006.2(DMD):c.3940C>T (p.Arg1314Ter)See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Becker muscular dystrophy by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the DMD gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00929] Limb-girdle muscular dystrophy

[00930] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Limb-girdle muscular dystrophy. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from SGCB, MYOT, LMNA, CAPN3, DYSF, SGCA, TTN, ANO5, TRAPPC11, LMNA, POMT1, and FKRP, including at least the followings:

NM_000232.4(SGCB):c.31C>T (p.Gln11Ter)

NM_006790.2(MYOT):c.164C>T (p.Ser55Phe)

NM_006790.2(MYOT):c.170C>T (p.Thr57Ile)

NM_170707.3(LMNA):c.1488+1G>A

NM_170707.3(LMNA):c.1609-1G>A

NM_000070.2(CAPN3):c.1715G>A (p.Arg572Gln)

NM_000070.2(CAPN3):c.2243G>A (p.Arg748Gln)

NM_000070.2(CAPN3):c.145C>T (p.Arg49Cys)

NM_000070.2(CAPN3):c.1319G>A (p.Arg440Gln)

NM_000070.2(CAPN3):c.1343G>A (p.Arg448His)

NM_000070.2(CAPN3):c.1465C>T (p.Arg489Trp)

NM_000070.2(CAPN3):c.1714C>T (p.Arg572Trp)

NM_000070.2(CAPN3):c.2306G>A (p.Arg769Gln)

NM_000070.2(CAPN3):c.133G>A (p.Ala45Thr)

NM_000070.2(CAPN3):c.499-1G>A

NM_000070.2(CAPN3):c.439C>T (p.Arg147Ter)

NM_000070.2(CAPN3):c.1063C>T (p.Arg355Trp)

NM_000070.2(CAPN3):c.1250C>T (p.Thr417Met)

NM_000070.2(CAPN3):c.245C>T (p.Pro82Leu)

NM_000070.2(CAPN3):c.2242C>T (p.Arg748Ter)

NM_000070.2(CAPN3):c.1318C>T (p.Arg440Trp)

NM_000070.2(CAPN3):c.1333G>A (p.Gly445Arg)

NM_000070.2(CAPN3):c.1957C>T (p.Gln653Ter)

NM_000070.2(CAPN3):c.1801-1G>A

NM_000070.2(CAPN3):c.2263+1G>A

NM_000070.2(CAPN3):c.956C>T (p.Pro319Leu)

NM_000070.2(CAPN3):c.1468C>T (p.Arg490Trp)

NM_000070.2(CAPN3):c.802-9G>A

NM_000070.2(CAPN3):c.1342C>T (p.Arg448Cys)

NM_000070.2(CAPN3):c.1303G>A (p.Glu435Lys)

NM_000070.2(CAPN3):c.1993-1G>A

NM_003494.3(DYSF):c.3113G>A (p.Arg1038Gln)

NM_001130987.1(DYSF):c.5174+1G>A

NM_001130987.1(DYSF):c.159G>A (p.Trp53Ter)

NM_001130987.1(DYSF):c.2929C>T (p.Arg977Trp)

NM_001130987.1(DYSF):c.4282C>T (p.Gln1428Ter)

NM_001130987.1(DYSF):c.1577-1G>A

NM_003494.3(DYSF):c.5529G>A (p.Trp1843Ter)

NM_001130987.1(DYSF):c.1576+1G>A

NM_001130987.1(DYSF):c.4462C>T (p.Gln1488Ter)

NM_003494.3(DYSF):c.5429G>A (p.Arg1810Lys)

NM_003494.3(DYSF):c.5077C>T (p.Arg1693Trp)

NM_001130978.1(DYSF):c.1813C>T (p.Gln605Ter)

NM_003494.3(DYSF):c.3230G>A (p.Trp1077Ter)

NM_003494.3(DYSF):c.265C>T (p.Arg89Ter)

NM_003494.3(DYSF):c.4434G>A (p.Trp1478Ter)

NM_003494.3(DYSF):c.3478C>T (p.Gln1160Ter)

NM_001130987.1(DYSF):c.1372G>A (p.Gly458Arg)

NM_003494.3(DYSF):c.4090C>T (p.Gln1364Ter)

NM_001130987.1(DYSF):c.2409+1G>A

NM_003494.3(DYSF):c.1708C>T (p.Gln570Ter)

NM_003494.3(DYSF):c.1956G>A (p.Trp652Ter)

NM_001130987.1(DYSF):c.5004-1G>A

NM_003494.3(DYSF):c.331C>T (p.Gln111Ter)

NM_001130978.1(DYSF):c.5776C>T (p.Arg1926Ter)

NM_003494.3(DYSF):c.6124C>T (p.Arg2042Cys)

NM_003494.3(DYSF):c.2643+1G>A

NM_003494.3(DYSF):c.4253G>A (p.Gly1418Asp)

NM_003494.3(DYSF):c.610C>T (p.Arg204Ter)

NM_003494.3(DYSF):c.1834C>T (p.Gln612Ter)

NM_003494.3(DYSF):c.5668-7G>A

NM_001130978.1(DYSF):c.3137G>A (p.Arg1046His)

NM_003494.3(DYSF):c.1053+1G>A

NM_003494.3(DYSF):c.1398-1G>A

NM_003494.3(DYSF):c.1481-1G>A

NM_003494.3(DYSF):c.2311C>T (p.Gln771Ter)

NM_003494.3(DYSF):c.2869C>T (p.Gln957Ter)

NM_003494.3(DYSF):c.4756C>T (p.Arg1586Ter)

NM_003494.3(DYSF):c.5509G>A (p.Asp1837Asn)

NM_003494.3(DYSF):c.5644C>T (p.Gln1882Ter)

NM_003494.3(DYSF):c.5946+1G>A

NM_003494.3(DYSF):c.937+1G>A

NM_003494.3(DYSF):c.5266C>T (p.Gln1756Ter)

NM_003494.3(DYSF):c.3832C>T (p.Gln1278Ter)

NM_003494.3(DYSF):c.5525+1G>A

NM_003494.3(DYSF):c.3112C>T (p.Arg1038Ter)

NM_000023.3(SGCA):c.293G>A (p.Arg98His)

NM_000023.3(SGCA):c.850C>T (p.Arg284Cys)

NM_000023.3(SGCA):c.403C>T (p.Gln135Ter)

NM_000023.3(SGCA):c.409G>A (p.Glu137Lys)

NM_000023.3(SGCA):c.747+1G>A

NM_000023.3(SGCA):c.229C>T (p.Arg77Cys)

NM_000023.3(SGCA):c.101G>A (p.Arg34His)

NM_000023.3(SGCA):c.739G>A (p.Val247Met)

NM_001256850.1(TTN):c.87394C>T (p.Arg29132Ter)

NM_213599.2(ANO5):c.762+1G>A

NM_213599.2(ANO5):c.1213C>T (p.Gln405Ter)

NM_213599.2(ANO5):c.1639C>T (p.Arg547Ter)

NM_213599.2(ANO5):c.1406G>A (p.Trp469Ter)

NM_213599.2(ANO5):c.1210C>T (p.Arg404Ter)

NM_213599.2(ANO5):c.2272C>T (p.Arg758Cys)

NM_213599.2(ANO5):c.41-1G>A

NM_213599.2(ANO5):c.172C>T (p.Arg58Trp)

NM_213599.2(ANO5):c.1898+1G>A

NM_021942.5(TRAPPC11):c.1287+5G>A

NM_170707.3(LMNA):c.1608+1G>A

NM_007171.3(POMT1):c.1864C>T (p.Arg622Ter)

NM_024301.4(FKRP):c.313C>T (p.Gln105Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Limb-girdle muscular dystrophy by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from SGCB, MYOT, LMNA, CAPN3, DYSF, SGCA, TTN, ANO5, TRAPPC11, LMNA, POMT1, and FKRP, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00931] Emery-Dreifuss muscular dystrophy

[00932] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Emery-Dreifuss muscular dystrophy. In some embodiment, the pathogenic mutations/SNPs are present in at least the EMD or SYNE1 gene, including at least the followings:

NM_000117.2(EMD):c.3G>A (p.Met1Ile)

NM_033071.3(SYNE1):c.11908C>T (p.Arg3970Ter)

NM_033071.3(SYNE1):c.21721C>T (p.Gln7241Ter)

NM_000117.2(EMD):c.130C>T (p.Gln44Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Emery-Dreifuss muscular dystrophy by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the EMD or SYNE1 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00933] Facioscapulohumeral muscular dystrophy

[00934] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Facioscapulohumeral muscular dystrophy. In some embodiment, the pathogenic mutations/SNPs are present in at least the SMCHD1 gene, including at least the followings:
NM_015295.2(SMCHD1):c.3801+1G>A
NM_015295.2(SMCHD1):c.1843-1G>A
See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Facioscapulohumeral muscular dystrophy by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the SMCHD1 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00935] **Inborn Errors of Metabolism (IEM)**

[00936] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various IEMs are reported in the ClinVar database and disclosed in Table A, including but not limited to Primary hyperoxaluria type 1, Argininosuccinate lyase deficiency, Ornithine carbamoyltransferase deficiency, and Maple syrup urine disease. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00937] Primary hyperoxaluria type 1

[00938] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Primary hyperoxaluria type 1. In some embodiment, the pathogenic mutations/SNPs are present in at least the AGXT gene, including at least the followings:
NM_000030.2(AGXT):c.245G>A (p.Gly82Glu)
NM_000030.2(AGXT):c.698G>A (p.Arg233His)
NM_000030.2(AGXT):c.466G>A (p.Gly156Arg)
NM_000030.2(AGXT):c.106C>T (p.Arg36Cys)
NM_000030.2(AGXT):c.346G>A (p.Gly116Arg)
NM_000030.2(AGXT):c.568G>A (p.Gly190Arg)
NM_000030.2(AGXT):c.653C>T (p.Ser218Leu)
NM_000030.2(AGXT):c.737G>A (p.Trp246Ter)
NM_000030.2(AGXT):c.1049G>A (p.Gly350Asp)
NM_000030.2(AGXT):c.473C>T (p.Ser158Leu)

NM_000030.2(AGXT):c.907C>T (p.Gln303Ter)

NM_000030.2(AGXT):c.996G>A (p.Trp332Ter)

NM_000030.2(AGXT):c.508G>A (p.Gly170Arg)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Primary hyperoxaluria type 1 by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the AGXT gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00939] Argininosuccinate lyase deficiency

[00940] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Argininosuccinate lyase deficiency. In some embodiment, the pathogenic mutations/SNPs are present in at least the ASL gene, including at least the followings:

NM_001024943.1(ASL):c.1153C>T (p.Arg385Cys)

NM_000048.3(ASL):c.532G>A (p.Val178Met)

NM_000048.3(ASL):c.545G>A (p.Arg182Gln)

NM_000048.3(ASL):c.175G>A (p.Glu59Lys)

NM_000048.3(ASL):c.718+5G>A

NM_000048.3(ASL):c.889C>T (p.Arg297Trp)

NM_000048.3(ASL):c.1360C>T (p.Gln454Ter)

NM_000048.3(ASL):c.1060C>T (p.Gln354Ter)

NM_000048.3(ASL):c.35G>A (p.Arg12Gln)

NM_000048.3(ASL):c.446+1G>A

NM_000048.3(ASL):c.544C>T (p.Arg182Ter)

NM_000048.3(ASL):c.1135C>T (p.Arg379Cys)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Argininosuccinate lyase deficiency by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the ASL gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00941] Ornithine carbamoyltransferase deficiency

[00942] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated

with Ornithine carbamoyltransferase deficiency. In some embodiment, the pathogenic mutations/SNPs are present in at least the OTC gene, including at least the followings:

NM_000531.5(OTC):c.119G>A (p.Arg40His)

NM_000531.5(OTC):c.422G>A (p.Arg141Gln)

NM_000531.5(OTC):c.829C>T (p.Arg277Trp)

NM_000531.5(OTC):c.674C>T (p.Pro225Leu)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Ornithine carbamoyltransferase deficiency by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the OTC gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00943] Maple syrup urine disease

[00944] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Maple syrup urine disease. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from BCKDHA, BCKDHB, DBT, and DLD, including at least the followings:

NM_000709.3(BCKDHA):c.476G>A (p.Arg159Gln)

NM_183050.3(BCKDHB):c.3G>A (p.Met1Ile)

NM_183050.3(BCKDHB):c.554C>T (p.Pro185Leu)

NM_001918.3(DBT):c.1033G>A (p.Gly345Arg)

NM_000709.3(BCKDHA):c.940C>T (p.Arg314Ter)

NM_000709.3(BCKDHA):c.793C>T (p.Arg265Trp)

NM_000709.3(BCKDHA):c.868G>A (p.Gly290Arg)

NM_000108.4(DLD):c.1123G>A (p.Glu375Lys)

NM_000709.3(BCKDHA):c.1234G>A (p.Val412Met)

NM_000709.3(BCKDHA):c.288+1G>A

NM_000709.3(BCKDHA):c.979G>A (p.Glu327Lys)

NM_001918.3(DBT):c.901C>T (p.Arg301Cys)

NM_183050.3(BCKDHB):c.509G>A (p.Arg170His)

NM_183050.3(BCKDHB):c.799C>T (p.Gln267Ter)

NM_183050.3(BCKDHB):c.853C>T (p.Arg285Ter)

NM_183050.3(BCKDHB):c.970C>T (p.Arg324Ter)

NM_183050.3(BCKDHB):c.832G>A (p.Gly278Ser)

NM_000709.3(BCKDHA):c.1036C>T (p.Arg346Cys)

NM_000709.3(BCKDHA):c.288+9C>T

NM_000709.3(BCKDHA):c.632C>T (p.Thr211Met)

NM_000709.3(BCKDHA):c.659C>T (p.Ala220Val)

NM_000709.3(BCKDHA):c.964C>T (p.Gln322Ter)

NM_001918.3(DBT):c.1291C>T (p.Arg431Ter)

NM_001918.3(DBT):c.251G>A (p.Trp84Ter)

NM_001918.3(DBT):c.871C>T (p.Arg291Ter)

NM_000056.4(BCKDHB):c.1016C>T (p.Ser339Leu)

NM_000056.4(BCKDHB):c.344-1G>A

NM_000056.4(BCKDHB):c.633+1G>A

NM_000056.4(BCKDHB):c.952-1G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Maple syrup urine disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from BCKDHA, BCKDHB, DBT, and DLD, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00945] Cancer-Related Diseases

[00946] Pathogenic G-to-A or C-to-T mutations/SNPs associated with various cancers and cancer-related diseases are reported in the ClinVar database and disclosed in Table A, including but not limited to Breast-Ovarian Cancer and Lynch syndrome. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00947] Breast-Ovarian Cancer

[00948] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Breast-Ovarian Cancer. In some embodiment, the pathogenic mutations/SNPs are present in at least the BRCA1or BRCA2 gene, including at least the followings:

NM_007294.3(BRCA1):c.5095C>T (p.Arg1699Trp)

NM_000059.3(BRCA2):c.7558C>T (p.Arg2520Ter)

NM_007294.3(BRCA1):c.2572C>T (p.Gln858Ter)

NM_007294.3(BRCA1):c.3607C>T (p.Arg1203Ter)

NM_007294.3(BRCA1):c.5503C>T (p.Arg1835Ter)

NM_007294.3(BRCA1):c.2059C>T (p.Gln687Ter)

NM_007294.3(BRCA1):c.4675+1G>A

NM_007294.3(BRCA1):c.5251C>T (p.Arg1751Ter)

NM_007294.3(BRCA1):c.5444G>A (p.Trp1815Ter)

NM_000059.3(BRCA2):c.9318G>A (p.Trp3106Ter)

NM_000059.3(BRCA2):c.9382C>T (p.Arg3128Ter)

NM_000059.3(BRCA2):c.274C>T (p.Gln92Ter)

NM_000059.3(BRCA2):c.6952C>T (p.Arg2318Ter)

NM_007294.3(BRCA1):c.1687C>T (p.Gln563Ter)

NM_007294.3(BRCA1):c.2599C>T (p.Gln867Ter)

NM_007294.3(BRCA1):c.784C>T (p.Gln262Ter)

NM_007294.3(BRCA1):c.280C>T (p.Gln94Ter)

NM_007294.3(BRCA1):c.5542C>T (p.Gln1848Ter)

NM_007294.3(BRCA1):c.5161C>T (p.Gln1721Ter)

NM_007294.3(BRCA1):c.4573C>T (p.Gln1525Ter)

NM_007294.3(BRCA1):c.4270C>T (p.Gln1424Ter)

NM_007294.3(BRCA1):c.4225C>T (p.Gln1409Ter)

NM_007294.3(BRCA1):c.4066C>T (p.Gln1356Ter)

NM_007294.3(BRCA1):c.3679C>T (p.Gln1227Ter)

NM_007294.3(BRCA1):c.1918C>T (p.Gln640Ter)

NM_007294.3(BRCA1):c.963G>A (p.Trp321Ter)

NM_007294.3(BRCA1):c.718C>T (p.Gln240Ter)

NM_000059.3(BRCA2):c.9196C>T (p.Gln3066Ter)

NM_000059.3(BRCA2):c.9154C>T (p.Arg3052Trp)

NM_007294.3(BRCA1):c.3991C>T (p.Gln1331Ter)

NM_007294.3(BRCA1):c.4097-1G>A

NM_007294.3(BRCA1):c.1059G>A (p.Trp353Ter)

NM_007294.3(BRCA1):c.1115G>A (p.Trp372Ter)

NM_007294.3(BRCA1):c.1138C>T (p.Gln380Ter)

NM_007294.3(BRCA1):c.1612C>T (p.Gln538Ter)

NM_007294.3(BRCA1):c.1621C>T (p.Gln541Ter)

NM_007294.3(BRCA1):c.1630C>T (p.Gln544Ter)

NM_007294.3(BRCA1):c.178C>T (p.Gln60Ter)

NM_007294.3(BRCA1):c.1969C>T (p.Gln657Ter)

NM_007294.3(BRCA1):c.2275C>T (p.Gln759Ter)

NM_007294.3(BRCA1):c.2410C>T (p.Gln804Ter)
NM_007294.3(BRCA1):c.2869C>T (p.Gln957Ter)
NM_007294.3(BRCA1):c.2923C>T (p.Gln975Ter)
NM_007294.3(BRCA1):c.3268C>T (p.Gln1090Ter)
NM_007294.3(BRCA1):c.3430C>T (p.Gln1144Ter)
NM_007294.3(BRCA1):c.3544C>T (p.Gln1182Ter)
NM_007294.3(BRCA1):c.4075C>T (p.Gln1359Ter)
NM_007294.3(BRCA1):c.4201C>T (p.Gln1401Ter)
NM_007294.3(BRCA1):c.4399C>T (p.Gln1467Ter)
NM_007294.3(BRCA1):c.4552C>T (p.Gln1518Ter)
NM_007294.3(BRCA1):c.5054C>T (p.Thr1685Ile)
NM_007294.3(BRCA1):c.514C>T (p.Gln172Ter)
NM_007294.3(BRCA1):c.5239C>T (p.Gln1747Ter)
NM_007294.3(BRCA1):c.5266C>T (p.Gln1756Ter)
NM_007294.3(BRCA1):c.5335C>T (p.Gln1779Ter)
NM_007294.3(BRCA1):c.5345G>A (p.Trp1782Ter)
NM_007294.3(BRCA1):c.5511G>A (p.Trp1837Ter)
NM_007294.3(BRCA1):c.5536C>T (p.Gln1846Ter)
NM_007294.3(BRCA1):c.55C>T (p.Gln19Ter)
NM_007294.3(BRCA1):c.949C>T (p.Gln317Ter)
NM_007294.3(BRCA1):c.928C>T (p.Gln310Ter)
NM_007294.3(BRCA1):c.5117G>A (p.Gly1706Glu)
NM_007294.3(BRCA1):c.5136G>A (p.Trp1712Ter)
NM_007294.3(BRCA1):c.4327C>T (p.Arg1443Ter)
NM_007294.3(BRCA1):c.1471C>T (p.Gln491Ter)
NM_007294.3(BRCA1):c.1576C>T (p.Gln526Ter)
NM_007294.3(BRCA1):c.160C>T (p.Gln54Ter)
NM_007294.3(BRCA1):c.2683C>T (p.Gln895Ter)
NM_007294.3(BRCA1):c.2761C>T (p.Gln921Ter)
NM_007294.3(BRCA1):c.3895C>T (p.Gln1299Ter)
NM_007294.3(BRCA1):c.4339C>T (p.Gln1447Ter)
NM_007294.3(BRCA1):c.4372C>T (p.Gln1458Ter)
NM_007294.3(BRCA1):c.5153G>A (p.Trp1718Ter)
NM_007294.3(BRCA1):c.5445G>A (p.Trp1815Ter)

NM_007294.3(BRCA1):c.5510G>A (p.Trp1837Ter)
NM_007294.3(BRCA1):c.5346G>A (p.Trp1782Ter)
NM_007294.3(BRCA1):c.1116G>A (p.Trp372Ter)
NM_007294.3(BRCA1):c.1999C>T (p.Gln667Ter)
NM_007294.3(BRCA1):c.4183C>T (p.Gln1395Ter)
NM_007294.3(BRCA1):c.4810C>T (p.Gln1604Ter)
NM_007294.3(BRCA1):c.850C>T (p.Gln284Ter)
NM_007294.3(BRCA1):c.1058G>A (p.Trp353Ter)
NM_007294.3(BRCA1):c.131G>A (p.Cys44Tyr)
NM_007294.3(BRCA1):c.1600C>T (p.Gln534Ter)
NM_007294.3(BRCA1):c.3286C>T (p.Gln1096Ter)
NM_007294.3(BRCA1):c.3403C>T (p.Gln1135Ter)
NM_007294.3(BRCA1):c.34C>T (p.Gln12Ter)
NM_007294.3(BRCA1):c.4258C>T (p.Gln1420Ter)
NM_007294.3(BRCA1):c.4609C>T (p.Gln1537Ter)
NM_007294.3(BRCA1):c.5154G>A (p.Trp1718Ter)
NM_007294.3(BRCA1):c.5431C>T (p.Gln1811Ter)
NM_007294.3(BRCA1):c.241C>T (p.Gln81Ter)
NM_007294.3(BRCA1):c.3331C>T (p.Gln1111Ter)
NM_007294.3(BRCA1):c.3967C>T (p.Gln1323Ter)
NM_007294.3(BRCA1):c.415C>T (p.Gln139Ter)
NM_007294.3(BRCA1):c.505C>T (p.Gln169Ter)
NM_007294.3(BRCA1):c.5194-12G>A
NM_007294.3(BRCA1):c.5212G>A (p.Gly1738Arg)
NM_007294.3(BRCA1):c.5332+1G>A
NM_007294.3(BRCA1):c.1480C>T (p.Gln494Ter)
NM_007294.3(BRCA1):c.2563C>T (p.Gln855Ter)
NM_007294.3(BRCA1):c.1066C>T (p.Gln356Ter)
NM_007294.3(BRCA1):c.3718C>T (p.Gln1240Ter)
NM_007294.3(BRCA1):c.3817C>T (p.Gln1273Ter)
NM_007294.3(BRCA1):c.3937C>T (p.Gln1313Ter)
NM_007294.3(BRCA1):c.4357+1G>A
NM_007294.3(BRCA1):c.5074+1G>A
NM_007294.3(BRCA1):c.5277+1G>A

NM_007294.3(BRCA1):c.2338C>T (p.Gln780Ter)
NM_007294.3(BRCA1):c.3598C>T (p.Gln1200Ter)
NM_007294.3(BRCA1):c.3841C>T (p.Gln1281Ter)
NM_007294.3(BRCA1):c.4222C>T (p.Gln1408Ter)
NM_007294.3(BRCA1):c.4524G>A (p.Trp1508Ter)
NM_007294.3(BRCA1):c.5353C>T (p.Gln1785Ter)
NM_007294.3(BRCA1):c.962G>A (p.Trp321Ter)
NM_007294.3(BRCA1):c.220C>T (p.Gln74Ter)
NM_007294.3(BRCA1):c.2713C>T (p.Gln905Ter)
NM_007294.3(BRCA1):c.2800C>T (p.Gln934Ter)
NM_007294.3(BRCA1):c.4612C>T (p.Gln1538Ter)
NM_007294.3(BRCA1):c.3352C>T (p.Gln1118Ter)
NM_007294.3(BRCA1):c.4834C>T (p.Gln1612Ter)
NM_007294.3(BRCA1):c.4523G>A (p.Trp1508Ter)
NM_007294.3(BRCA1):c.5135G>A (p.Trp1712Ter)
NM_007294.3(BRCA1):c.1155G>A (p.Trp385Ter)
NM_007294.3(BRCA1):c.4987-1G>A
NM_000059.3(BRCA2):c.9573G>A (p.Trp3191Ter)
NM_000059.3(BRCA2):c.1945C>T (p.Gln649Ter)
NM_000059.3(BRCA2):c.217C>T (p.Gln73Ter)
NM_000059.3(BRCA2):c.523C>T (p.Gln175Ter)
NM_000059.3(BRCA2):c.2548C>T (p.Gln850Ter)
NM_000059.3(BRCA2):c.2905C>T (p.Gln969Ter)
NM_000059.3(BRCA2):c.4689G>A (p.Trp1563Ter)
NM_000059.3(BRCA2):c.4972C>T (p.Gln1658Ter)
NM_000059.3(BRCA2):c.1184G>A (p.Trp395Ter)
NM_000059.3(BRCA2):c.2137C>T (p.Gln713Ter)
NM_000059.3(BRCA2):c.3217C>T (p.Gln1073Ter)
NM_000059.3(BRCA2):c.3523C>T (p.Gln1175Ter)
NM_000059.3(BRCA2):c.4783C>T (p.Gln1595Ter)
NM_000059.3(BRCA2):c.5800C>T (p.Gln1934Ter)
NM_000059.3(BRCA2):c.6478C>T (p.Gln2160Ter)
NM_000059.3(BRCA2):c.7033C>T (p.Gln2345Ter)
NM_000059.3(BRCA2):c.7495C>T (p.Gln2499Ter)

NM_000059.3(BRCA2):c.7501C>T (p.Gln2501Ter)
NM_000059.3(BRCA2):c.7887G>A (p.Trp2629Ter)
NM_000059.3(BRCA2):c.8910G>A (p.Trp2970Ter)
NM_000059.3(BRCA2):c.9139C>T (p.Gln3047Ter)
NM_000059.3(BRCA2):c.9739C>T (p.Gln3247Ter)
NM_000059.3(BRCA2):c.582G>A (p.Trp194Ter)
NM_000059.3(BRCA2):c.7963C>T (p.Gln2655Ter)
NM_000059.3(BRCA2):c.8695C>T (p.Gln2899Ter)
NM_000059.3(BRCA2):c.8869C>T (p.Gln2957Ter)
NM_000059.3(BRCA2):c.1117C>T (p.Gln373Ter)
NM_000059.3(BRCA2):c.1825C>T (p.Gln609Ter)
NM_000059.3(BRCA2):c.2455C>T (p.Gln819Ter)
NM_000059.3(BRCA2):c.2881C>T (p.Gln961Ter)
NM_000059.3(BRCA2):c.3265C>T (p.Gln1089Ter)
NM_000059.3(BRCA2):c.3283C>T (p.Gln1095Ter)
NM_000059.3(BRCA2):c.3442C>T (p.Gln1148Ter)
NM_000059.3(BRCA2):c.3871C>T (p.Gln1291Ter)
NM_000059.3(BRCA2):c.439C>T (p.Gln147Ter)
NM_000059.3(BRCA2):c.4525C>T (p.Gln1509Ter)
NM_000059.3(BRCA2):c.475+1G>A
NM_000059.3(BRCA2):c.5344C>T (p.Gln1782Ter)
NM_000059.3(BRCA2):c.5404C>T (p.Gln1802Ter)
NM_000059.3(BRCA2):c.5773C>T (p.Gln1925Ter)
NM_000059.3(BRCA2):c.5992C>T (p.Gln1998Ter)
NM_000059.3(BRCA2):c.6469C>T (p.Gln2157Ter)
NM_000059.3(BRCA2):c.7261C>T (p.Gln2421Ter)
NM_000059.3(BRCA2):c.7303C>T (p.Gln2435Ter)
NM_000059.3(BRCA2):c.7471C>T (p.Gln2491Ter)
NM_000059.3(BRCA2):c.7681C>T (p.Gln2561Ter)
NM_000059.3(BRCA2):c.7738C>T (p.Gln2580Ter)
NM_000059.3(BRCA2):c.7886G>A (p.Trp2629Ter)
NM_000059.3(BRCA2):c.8140C>T (p.Gln2714Ter)
NM_000059.3(BRCA2):c.8363G>A (p.Trp2788Ter)
NM_000059.3(BRCA2):c.8572C>T (p.Gln2858Ter)

NM_000059.3(BRCA2):c.8773C>T (p.Gln2925Ter)
NM_000059.3(BRCA2):c.8821C>T (p.Gln2941Ter)
NM_000059.3(BRCA2):c.9109C>T (p.Gln3037Ter)
NM_000059.3(BRCA2):c.9317G>A (p.Trp3106Ter)
NM_000059.3(BRCA2):c.9466C>T (p.Gln3156Ter)
NM_000059.3(BRCA2):c.9572G>A (p.Trp3191Ter)
NM_000059.3(BRCA2):c.8490G>A (p.Trp2830Ter)
NM_000059.3(BRCA2):c.5980C>T (p.Gln1994Ter)
NM_000059.3(BRCA2):c.7721G>A (p.Trp2574Ter)
NM_000059.3(BRCA2):c.196C>T (p.Gln66Ter)
NM_000059.3(BRCA2):c.7618-1G>A
NM_000059.3(BRCA2):c.8489G>A (p.Trp2830Ter)
NM_000059.3(BRCA2):c.7857G>A (p.Trp2619Ter)
NM_000059.3(BRCA2):c.1261C>T (p.Gln421Ter)
NM_000059.3(BRCA2):c.1456C>T (p.Gln486Ter)
NM_000059.3(BRCA2):c.3319C>T (p.Gln1107Ter)
NM_000059.3(BRCA2):c.5791C>T (p.Gln1931Ter)
NM_000059.3(BRCA2):c.6070C>T (p.Gln2024Ter)
NM_000059.3(BRCA2):c.7024C>T (p.Gln2342Ter)
NM_000059.3(BRCA2):c.961C>T (p.Gln321Ter)
NM_000059.3(BRCA2):c.9380G>A (p.Trp3127Ter)
NM_000059.3(BRCA2):c.8364G>A (p.Trp2788Ter)
NM_000059.3(BRCA2):c.7758G>A (p.Trp2586Ter)
NM_000059.3(BRCA2):c.2224C>T (p.Gln742Ter)
NM_000059.3(BRCA2):c.5101C>T (p.Gln1701Ter)
NM_000059.3(BRCA2):c.5959C>T (p.Gln1987Ter)
NM_000059.3(BRCA2):c.7060C>T (p.Gln2354Ter)
NM_000059.3(BRCA2):c.9100C>T (p.Gln3034Ter)
NM_000059.3(BRCA2):c.9148C>T (p.Gln3050Ter)
NM_000059.3(BRCA2):c.9883C>T (p.Gln3295Ter)
NM_000059.3(BRCA2):c.1414C>T (p.Gln472Ter)
NM_000059.3(BRCA2):c.1689G>A (p.Trp563Ter)
NM_000059.3(BRCA2):c.581G>A (p.Trp194Ter)
NM_000059.3(BRCA2):c.6490C>T (p.Gln2164Ter)

NM_000059.3(BRCA2):c.7856G>A (p.Trp2619Ter)
NM_000059.3(BRCA2):c.8970G>A (p.Trp2990Ter)
NM_000059.3(BRCA2):c.92G>A (p.Trp31Ter)
NM_000059.3(BRCA2):c.9376C>T (p.Gln3126Ter)
NM_000059.3(BRCA2):c.93G>A (p.Trp31Ter)
NM_000059.3(BRCA2):c.1189C>T (p.Gln397Ter)
NM_000059.3(BRCA2):c.2818C>T (p.Gln940Ter)
NM_000059.3(BRCA2):c.2979G>A (p.Trp993Ter)
NM_000059.3(BRCA2):c.3166C>T (p.Gln1056Ter)
NM_000059.3(BRCA2):c.4285C>T (p.Gln1429Ter)
NM_000059.3(BRCA2):c.6025C>T (p.Gln2009Ter)
NM_000059.3(BRCA2):c.772C>T (p.Gln258Ter)
NM_000059.3(BRCA2):c.7877G>A (p.Trp2626Ter)
NM_000059.3(BRCA2):c.3109C>T (p.Gln1037Ter)
NM_000059.3(BRCA2):c.4222C>T (p.Gln1408Ter)
NM_000059.3(BRCA2):c.7480C>T (p.Arg2494Ter)
NM_000059.3(BRCA2):c.7878G>A (p.Trp2626Ter)
NM_000059.3(BRCA2):c.9076C>T (p.Gln3026Ter)
NM_000059.3(BRCA2):c.1855C>T (p.Gln619Ter)
NM_000059.3(BRCA2):c.4111C>T (p.Gln1371Ter)
NM_000059.3(BRCA2):c.5656C>T (p.Gln1886Ter)
NM_000059.3(BRCA2):c.7757G>A (p.Trp2586Ter)
NM_000059.3(BRCA2):c.8243G>A (p.Gly2748Asp)
NM_000059.3(BRCA2):c.8878C>T (p.Gln2960Ter)
NM_000059.3(BRCA2):c.8487+1G>A
NM_000059.3(BRCA2):c.8677C>T (p.Gln2893Ter)
NM_000059.3(BRCA2):c.250C>T (p.Gln84Ter)
NM_000059.3(BRCA2):c.6124C>T (p.Gln2042Ter)
NM_000059.3(BRCA2):c.7617+1G>A
NM_000059.3(BRCA2):c.8575C>T (p.Gln2859Ter)
NM_000059.3(BRCA2):c.8174G>A (p.Trp2725Ter)
NM_000059.3(BRCA2):c.3187C>T (p.Gln1063Ter)
NM_000059.3(BRCA2):c.9381G>A (p.Trp3127Ter)
NM_000059.3(BRCA2):c.2095C>T (p.Gln699Ter)

NM_000059.3(BRCA2):c.1642C>T (p.Gln548Ter)

NM_000059.3(BRCA2):c.8608C>T (p.Gln2870Ter)

NM_000059.3(BRCA2):c.3412C>T (p.Gln1138Ter)

NM_000059.3(BRCA2):c.4246C>T (p.Gln1416Ter)

NM_000059.3(BRCA2):c.6475C>T (p.Gln2159Ter)

NM_000059.3(BRCA2):c.7366C>T (p.Gln2456Ter)

NM_000059.3(BRCA2):c.7516C>T (p.Gln2506Ter)

NM_000059.3(BRCA2):c.8969G>A (p.Trp2990Ter)

NM_000059.3(BRCA2):c.6487C>T (p.Gln2163Ter)

NM_000059.3(BRCA2):c.2978G>A (p.Trp993Ter)

NM_000059.3(BRCA2):c.7615C>T (p.Gln2539Ter)

NM_000059.3(BRCA2):c.9106C>T (p.Gln3036Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Breast-Ovarian Cancer by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the BRCA1or BRCA2 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00949] Lynch syndrome

[00950] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Lynch syndrome. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from MSH6, MSH2, EPCAM, PMS2, and MLH1, including at least the followings:

NM_000179.2(MSH6):c.1045C>T (p.Gln349Ter)

NM_000251.2(MSH2):c.1384C>T (p.Gln462Ter)

NM_002354.2(EPCAM):c.133C>T (p.Gln45Ter)

NM_002354.2(EPCAM):c.429G>A (p.Trp143Ter)

NM_002354.2(EPCAM):c.523C>T (p.Gln175Ter)

NM_000179.2(MSH6):c.2680C>T (p.Gln894Ter)

NM_000251.2(MSH2):c.350G>A (p.Trp117Ter)

NM_000179.2(MSH6):c.2735G>A (p.Trp912Ter)

NM_000179.2(MSH6):c.3556+1G>A

NM_000251.2(MSH2):c.388C>T (p.Gln130Ter)

NM_000535.6(PMS2):c.1912C>T (p.Gln638Ter)

NM_000535.6(PMS2):c.1891C>T (p.Gln631Ter)
NM_000249.3(MLH1):c.454-1G>A
NM_000251.2(MSH2):c.1030C>T (p.Gln344Ter)
NM_000179.2(MSH6):c.2330G>A (p.Trp777Ter)
NM_000179.2(MSH6):c.2191C>T (p.Gln731Ter)
NM_000179.2(MSH6):c.2764C>T (p.Arg922Ter)
NM_000179.2(MSH6):c.2815C>T (p.Gln939Ter)
NM_000179.2(MSH6):c.3020G>A (p.Trp1007Ter)
NM_000179.2(MSH6):c.3436C>T (p.Gln1146Ter)
NM_000179.2(MSH6):c.3647-1G>A
NM_000179.2(MSH6):c.3772C>T (p.Gln1258Ter)
NM_000179.2(MSH6):c.3838C>T (p.Gln1280Ter)
NM_000179.2(MSH6):c.706C>T (p.Gln236Ter)
NM_000179.2(MSH6):c.730C>T (p.Gln244Ter)
NM_000249.3(MLH1):c.1171C>T (p.Gln391Ter)
NM_000249.3(MLH1):c.1192C>T (p.Gln398Ter)
NM_000249.3(MLH1):c.1225C>T (p.Gln409Ter)
NM_000249.3(MLH1):c.1276C>T (p.Gln426Ter)
NM_000249.3(MLH1):c.1528C>T (p.Gln510Ter)
NM_000249.3(MLH1):c.1609C>T (p.Gln537Ter)
NM_000249.3(MLH1):c.1613G>A (p.Trp538Ter)
NM_000249.3(MLH1):c.1614G>A (p.Trp538Ter)
NM_000249.3(MLH1):c.1624C>T (p.Gln542Ter)
NM_000249.3(MLH1):c.1684C>T (p.Gln562Ter)
NM_000249.3(MLH1):c.1731+1G>A
NM_000249.3(MLH1):c.1731+5G>A
NM_000249.3(MLH1):c.1732-1G>A
NM_000249.3(MLH1):c.1896G>A (p.Glu632=)
NM_000249.3(MLH1):c.1989+1G>A
NM_000249.3(MLH1):c.1990-1G>A
NM_000249.3(MLH1):c.1998G>A (p.Trp666Ter)
NM_000249.3(MLH1):c.208-1G>A
NM_000249.3(MLH1):c.2101C>T (p.Gln701Ter)
NM_000249.3(MLH1):c.2136G>A (p.Trp712Ter)

NM_000249.3(MLH1):c.2224C>T (p.Gln742Ter)
NM_000249.3(MLH1):c.230G>A (p.Cys77Tyr)
NM_000249.3(MLH1):c.256C>T (p.Gln86Ter)
NM_000249.3(MLH1):c.436C>T (p.Gln146Ter)
NM_000249.3(MLH1):c.445C>T (p.Gln149Ter)
NM_000249.3(MLH1):c.545G>A (p.Arg182Lys)
NM_000249.3(MLH1):c.731G>A (p.Gly244Asp)
NM_000249.3(MLH1):c.76C>T (p.Gln26Ter)
NM_000249.3(MLH1):c.842C>T (p.Ala281Val)
NM_000249.3(MLH1):c.882C>T (p.Leu294=)
NM_000249.3(MLH1):c.901C>T (p.Gln301Ter)
NM_000251.2(MSH2):c.1013G>A (p.Gly338Glu)
NM_000251.2(MSH2):c.1034G>A (p.Trp345Ter)
NM_000251.2(MSH2):c.1129C>T (p.Gln377Ter)
NM_000251.2(MSH2):c.1183C>T (p.Gln395Ter)
NM_000251.2(MSH2):c.1189C>T (p.Gln397Ter)
NM_000251.2(MSH2):c.1204C>T (p.Gln402Ter)
NM_000251.2(MSH2):c.1276+1G>A
NM_000251.2(MSH2):c.1528C>T (p.Gln510Ter)
NM_000251.2(MSH2):c.1552C>T (p.Gln518Ter)
NM_000251.2(MSH2):c.1720C>T (p.Gln574Ter)
NM_000251.2(MSH2):c.1777C>T (p.Gln593Ter)
NM_000251.2(MSH2):c.1885C>T (p.Gln629Ter)
NM_000251.2(MSH2):c.2087C>T (p.Pro696Leu)
NM_000251.2(MSH2):c.2251G>A (p.Gly751Arg)
NM_000251.2(MSH2):c.2291G>A (p.Trp764Ter)
NM_000251.2(MSH2):c.2292G>A (p.Trp764Ter)
NM_000251.2(MSH2):c.2446C>T (p.Gln816Ter)
NM_000251.2(MSH2):c.2470C>T (p.Gln824Ter)
NM_000251.2(MSH2):c.2536C>T (p.Gln846Ter)
NM_000251.2(MSH2):c.2581C>T (p.Gln861Ter)
NM_000251.2(MSH2):c.2634G>A (p.Glu878=)
NM_000251.2(MSH2):c.2635C>T (p.Gln879Ter)
NM_000251.2(MSH2):c.28C>T (p.Gln10Ter)

NM_000251.2(MSH2):c.472C>T (p.Gln158Ter)
NM_000251.2(MSH2):c.478C>T (p.Gln160Ter)
NM_000251.2(MSH2):c.484G>A (p.Gly162Arg)
NM_000251.2(MSH2):c.490G>A (p.Gly164Arg)
NM_000251.2(MSH2):c.547C>T (p.Gln183Ter)
NM_000251.2(MSH2):c.577C>T (p.Gln193Ter)
NM_000251.2(MSH2):c.643C>T (p.Gln215Ter)
NM_000251.2(MSH2):c.645+1G>A
NM_000251.2(MSH2):c.652C>T (p.Gln218Ter)
NM_000251.2(MSH2):c.754C>T (p.Gln252Ter)
NM_000251.2(MSH2):c.792+1G>A
NM_000251.2(MSH2):c.942G>A (p.Gln314=)
NM_000535.6(PMS2):c.949C>T (p.Gln317Ter)
NM_000249.3(MLH1):c.306+1G>A
NM_000249.3(MLH1):c.62C>T (p.Ala21Val)
NM_000251.2(MSH2):c.1865C>T (p.Pro622Leu)
NM_000179.2(MSH6):c.426G>A (p.Trp142Ter)
NM_000251.2(MSH2):c.715C>T (p.Gln239Ter)
NM_000249.3(MLH1):c.350C>T (p.Thr117Met)
NM_000251.2(MSH2):c.1915C>T (p.His639Tyr)
NM_000251.2(MSH2):c.289C>T (p.Gln97Ter)
NM_000251.2(MSH2):c.2785C>T (p.Arg929Ter)
NM_000249.3(MLH1):c.131C>T (p.Ser44Phe)
NM_000249.3(MLH1):c.1219C>T (p.Gln407Ter)
NM_000249.3(MLH1):c.306+5G>A
NM_000251.2(MSH2):c.1801C>T (p.Gln601Ter)
NM_000535.6(PMS2):c.1144+1G>A
NM_000251.2(MSH2):c.1984C>T (p.Gln662Ter)
NM_000249.3(MLH1):c.381-1G>A
NM_000535.6(PMS2):c.631C>T (p.Arg211Ter)
NM_000251.2(MSH2):c.790C>T (p.Gln264Ter)
NM_000251.2(MSH2):c.366+1G>A
NM_000249.3(MLH1):c.298C>T (p.Arg100Ter)
NM_000179.2(MSH6):c.3013C>T (p.Arg1005Ter)

NM_000179.2(MSH6):c.694C>T (p.Gln232Ter)

NM_000179.2(MSH6):c.742C>T (p.Arg248Ter)

NM_000249.3(MLH1):c.1039-1G>A

NM_000249.3(MLH1):c.142C>T (p.Gln48Ter)

NM_000249.3(MLH1):c.1790G>A (p.Trp597Ter)

NM_000249.3(MLH1):c.1961C>T (p.Pro654Leu)

NM_000249.3(MLH1):c.2103+1G>A

NM_000249.3(MLH1):c.2135G>A (p.Trp712Ter)

NM_000249.3(MLH1):c.588+5G>A

NM_000249.3(MLH1):c.790+1G>A

NM_000251.2(MSH2):c.1035G>A (p.Trp345Ter)

NM_000251.2(MSH2):c.1255C>T (p.Gln419Ter)

NM_000251.2(MSH2):c.1861C>T (p.Arg621Ter)

NM_000251.2(MSH2):c.226C>T (p.Gln76Ter)

NM_000251.2(MSH2):c.2653C>T (p.Gln885Ter)

NM_000251.2(MSH2):c.508C>T (p.Gln170Ter)

NM_000251.2(MSH2):c.862C>T (p.Gln288Ter)

NM_000251.2(MSH2):c.892C>T (p.Gln298Ter)

NM_000251.2(MSH2):c.970C>T (p.Gln324Ter)

NM_000179.2(MSH6):c.4001G>A (p.Arg1334Gln)

NM_000251.2(MSH2):c.1662-1G>A

NM_000535.6(PMS2):c.1882C>T (p.Arg628Ter)

NM_000535.6(PMS2):c.2174+1G>A

NM_000535.6(PMS2):c.2404C>T (p.Arg802Ter)

NM_000179.2(MSH6):c.3991C>T (p.Arg1331Ter)

NM_000179.2(MSH6):c.2503C>T (p.Gln835Ter)

NM_000179.2(MSH6):c.718C>T (p.Arg240Ter)

NM_000249.3(MLH1):c.1038G>A (p.Gln346=)

NM_000249.3(MLH1):c.245C>T (p.Thr82Ile)

NM_000249.3(MLH1):c.83C>T (p.Pro28Leu)

NM_000249.3(MLH1):c.884G>A (p.Ser295Asn)

NM_000249.3(MLH1):c.982C>T (p.Gln328Ter)

NM_000251.2(MSH2):c.1046C>T (p.Pro349Leu)

NM_000251.2(MSH2):c.1120C>T (p.Gln374Ter)

NM_000251.2(MSH2):c.1285C>T (p.Gln429Ter)
NM_000251.2(MSH2):c.1477C>T (p.Gln493Ter)
NM_000251.2(MSH2):c.2152C>T (p.Gln718Ter)
NM_000535.6(PMS2):c.703C>T (p.Gln235Ter)
NM_000249.3(MLH1):c.2141G>A (p.Trp714Ter)
NM_000251.2(MSH2):c.1009C>T (p.Gln337Ter)
NM_000251.2(MSH2):c.1216C>T (p.Arg406Ter)
NM_000179.2(MSH6):c.3202C>T (p.Arg1068Ter)
NM_000251.2(MSH2):c.1165C>T (p.Arg389Ter)
NM_000249.3(MLH1):c.1943C>T (p.Pro648Leu)
NM_000249.3(MLH1):c.200G>A (p.Gly67Glu)
NM_000249.3(MLH1):c.793C>T (p.Arg265Cys)
NM_000249.3(MLH1):c.2059C>T (p.Arg687Trp)
NM_000249.3(MLH1):c.677G>A (p.Arg226Gln)
NM_000249.3(MLH1):c.2041G>A (p.Ala681Thr)
NM_000249.3(MLH1):c.1942C>T (p.Pro648Ser)
NM_000249.3(MLH1):c.676C>T (p.Arg226Ter)
NM_000251.2(MSH2):c.2038C>T (p.Arg680Ter)
NM_000179.2(MSH6):c.1483C>T (p.Arg495Ter)
NM_000179.2(MSH6):c.2194C>T (p.Arg732Ter)
NM_000179.2(MSH6):c.3103C>T (p.Arg1035Ter)
NM_000179.2(MSH6):c.892C>T (p.Arg298Ter)
NM_000249.3(MLH1):c.1459C>T (p.Arg487Ter)
NM_000249.3(MLH1):c.1731G>A (p.Ser577=)
NM_000249.3(MLH1):c.184C>T (p.Gln62Ter)
NM_000249.3(MLH1):c.1975C>T (p.Arg659Ter)
NM_000249.3(MLH1):c.199G>A (p.Gly67Arg)
NM_000251.2(MSH2):c.1076+1G>A
NM_000251.2(MSH2):c.1147C>T (p.Arg383Ter)
NM_000251.2(MSH2):c.181C>T (p.Gln61Ter)
NM_000251.2(MSH2):c.212-1G>A
NM_000251.2(MSH2):c.2131C>T (p.Arg711Ter)
NM_000535.6(PMS2):c.697C>T (p.Gln233Ter)
NM_000535.6(PMS2):c.1261C>T (p.Arg421Ter)

NM_000251.2(MSH2):c.2047G>A (p.Gly683Arg)

NM_000535.6(PMS2):c.400C>T (p.Arg134Ter)

NM_000535.6(PMS2):c.1927C>T (p.Gln643Ter)

NM_000179.2(MSH6):c.1444C>T (p.Arg482Ter)

NM_000179.2(MSH6):c.2731C>T (p.Arg911Ter)

NM_000535.6(PMS2):c.943C>T (p.Arg315Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Lynch syndrome by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from BCKDHA, BCKDHB, DBT, and DLD, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00951] Other Genetic Diseases

[00952] Pathogenic G-to-A or C-to-T mutations/SNPs associated with additional genetic diseases are also reported in the ClinVar database and disclosed in Table A, including but not limited to Marfan syndrome, Hurler syndrome, Glycogen Storage Disease, and Cystic Fibrosis. Accordingly, an aspect of the invention relates to a method for correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with any of these diseases, as discussed below.

[00953] Marfan syndrome

[00954] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Marfan syndrome. In some embodiment, the pathogenic mutations/SNPs are present in at least the FBN1 gene, including at least the followings:

NM_000138.4(FBN1):c.1879C>T (p.Arg627Cys)

NM_000138.4(FBN1):c.1051C>T (p.Gln351Ter)

NM_000138.4(FBN1):c.184C>T (p.Arg62Cys)

NM_000138.4(FBN1):c.2855-1G>A

NM_000138.4(FBN1):c.3164G>A (p.Cys1055Tyr)

NM_000138.4(FBN1):c.368G>A (p.Cys123Tyr)

NM_000138.4(FBN1):c.4955G>A (p.Cys1652Tyr)

NM_000138.4(FBN1):c.7180C>T (p.Arg2394Ter)

NM_000138.4(FBN1):c.8267G>A (p.Trp2756Ter)

NM_000138.4(FBN1):c.1496G>A (p.Cys499Tyr)

NM_000138.4(FBN1):c.6886C>T (p.Gln2296Ter)

NM_000138.4(FBN1):c.3373C>T (p.Arg1125Ter)
NM_000138.4(FBN1):c.640G>A (p.Gly214Ser)
NM_000138.4(FBN1):c.5038C>T (p.Gln1680Ter)
NM_000138.4(FBN1):c.434G>A (p.Cys145Tyr)
NM_000138.4(FBN1):c.2563C>T (p.Gln855Ter)
NM_000138.4(FBN1):c.7466G>A (p.Cys2489Tyr)
NM_000138.4(FBN1):c.2089C>T (p.Gln697Ter)
NM_000138.4(FBN1):c.592C>T (p.Gln198Ter)
NM_000138.4(FBN1):c.6695G>A (p.Cys2232Tyr)
NM_000138.4(FBN1):c.6164-1G>A
NM_000138.4(FBN1):c.5627G>A (p.Cys1876Tyr)
NM_000138.4(FBN1):c.4061G>A (p.Trp1354Ter)
NM_000138.4(FBN1):c.1982G>A (p.Cys661Tyr)
NM_000138.4(FBN1):c.6784C>T (p.Gln2262Ter)
NM_000138.4(FBN1):c.409C>T (p.Gln137Ter)
NM_000138.4(FBN1):c.364C>T (p.Arg122Cys)
NM_000138.4(FBN1):c.3217G>A (p.Glu1073Lys)
NM_000138.4(FBN1):c.4460-8G>A
NM_000138.4(FBN1):c.4786C>T (p.Arg1596Ter)
NM_000138.4(FBN1):c.7806G>A (p.Trp2602Ter)
NM_000138.4(FBN1):c.247+1G>A
NM_000138.4(FBN1):c.2495G>A (p.Cys832Tyr)
NM_000138.4(FBN1):c.493C>T (p.Arg165Ter)
NM_000138.4(FBN1):c.5504G>A (p.Cys1835Tyr)
NM_000138.4(FBN1):c.5863C>T (p.Gln1955Ter)
NM_000138.4(FBN1):c.6658C>T (p.Arg2220Ter)
NM_000138.4(FBN1):c.7606G>A (p.Gly2536Arg)
NM_000138.4(FBN1):c.7955G>A (p.Cys2652Tyr)
NM_000138.4(FBN1):c.3037G>A (p.Gly1013Arg)
NM_000138.4(FBN1):c.8080C>T (p.Arg2694Ter)
NM_000138.4(FBN1):c.1633C>T (p.Arg545Cys)
NM_000138.4(FBN1):c.7205-1G>A
NM_000138.4(FBN1):c.4621C>T (p.Arg1541Ter)
NM_000138.4(FBN1):c.1090C>T (p.Arg364Ter)

NM_000138.4(FBN1):c.1585C>T (p.Arg529Ter)

NM_000138.4(FBN1):c.4781G>A (p.Gly1594Asp)

NM_000138.4(FBN1):c.643C>T (p.Arg215Ter)

NM_000138.4(FBN1):c.3668G>A (p.Cys1223Tyr)

NM_000138.4(FBN1):c.8326C>T (p.Arg2776Ter)

NM_000138.4(FBN1):c.6354C>T (p.Ile2118=)

NM_000138.4(FBN1):c.1468+5G>A

NM_000138.4(FBN1):c.1546C>T (p.Arg516Ter)

NM_000138.4(FBN1):c.4615C>T (p.Arg1539Ter)

NM_000138.4(FBN1):c.5368C>T (p.Arg1790Ter)

NM_000138.4(FBN1):c.1285C>T (p.Arg429Ter)

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Marfan syndrome by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the FBN1 gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00955] Hurler syndrome

[00956] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Hurler syndrome. In some embodiment, the pathogenic mutations/SNPs are present in at least the IDUA gene, including at least the followings:

NM_000203.4(IDUA):c.972+1G>A

NM_000203.4(IDUA):c.1855C>T (p.Arg619Ter)

NM_000203.4(IDUA):c.152G>A (p.Gly51Asp)

NM_000203.4(IDUA):c.1205G>A (p.Trp402Ter)

NM_000203.4(IDUA):c.208C>T (p.Gln70Ter)

NM_000203.4(IDUA):c.1045G>A (p.Asp349Asn)

NM_000203.4(IDUA):c.1650+5G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Hurler syndrome by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the IDUA gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00957] Glycogen Storage Disease

[00958] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Glycogen Storage Disease. In some embodiment, the pathogenic mutations/SNPs are present in at least one gene selected from GAA, AGL, PHKB, PRKAG2, G6PC, PGAM2, GBE1, PYGM, and PFKM, including at least the followings:

NM_000152.4(GAA):c.1927G>A (p.Gly643Arg)

NM_000152.4(GAA):c.2173C>T (p.Arg725Trp)

NM_000642.2(AGL):c.3980G>A (p.Trp1327Ter)

NM_000642.2(AGL):c.16C>T (p.Gln6Ter)

NM_000642.2(AGL):c.2039G>A (p.Trp680Ter)

NM_000293.2(PHKB):c.1546C>T (p.Gln516Ter)

NM_016203.3(PRKAG2):c.1592G>A (p.Arg531Gln)

NM_000151.3(G6PC):c.248G>A (p.Arg83His)

NM_000151.3(G6PC):c.724C>T (p.Gln242Ter)

NM_000151.3(G6PC):c.883C>T (p.Arg295Cys)

NM_000151.3(G6PC):c.247C>T (p.Arg83Cys)

NM_000151.3(G6PC):c.1039C>T (p.Gln347Ter)

NM_000152.4(GAA):c.1561G>A (p.Glu521Lys)

NM_000642.2(AGL):c.2590C>T (p.Arg864Ter)

NM_000642.2(AGL):c.3682C>T (p.Arg1228Ter)

NM_000642.2(AGL):c.118C>T (p.Gln40Ter)

NM_000642.2(AGL):c.256C>T (p.Gln86Ter)

NM_000642.2(AGL):c.2681+1G>A

NM_000642.2(AGL):c.2158-1G>A

NM_000290.3(PGAM2):c.233G>A (p.Trp78Ter)

NM_000152.4(GAA):c.1548G>A (p.Trp516Ter)

NM_000152.4(GAA):c.2014C>T (p.Arg672Trp)

NM_000152.4(GAA):c.546G>A (p.Thr182=)

NM_000152.4(GAA):c.1802C>T (p.Ser601Leu)

NM_000152.4(GAA):c.1754+1G>A

NM_000152.4(GAA):c.1082C>T (p.Pro361Leu)

NM_000152.4(GAA):c.2560C>T (p.Arg854Ter)

NM_000152.4(GAA):c.655G>A (p.Gly219Arg)

NM_000152.4(GAA):c.1933G>A (p.Asp645Asn)

NM_000152.4(GAA):c.1979G>A (p.Arg660His)

NM_000152.4(GAA):c.1465G>A (p.Asp489Asn)

NM_000152.4(GAA):c.2512C>T (p.Gln838Ter)

NM_000158.3(GBE1):c.1543C>T (p.Arg515Cys)

NM_005609.3(PYGM):c.1726C>T (p.Arg576Ter)

NM_005609.3(PYGM):c.1827G>A (p.Lys609=)

NM_005609.3(PYGM):c.148C>T (p.Arg50Ter)

NM_005609.3(PYGM):c.613G>A (p.Gly205Ser)

NM_005609.3(PYGM):c.1366G>A (p.Val456Met)

NM_005609.3(PYGM):c.1768+1G>A

NM_001166686.1(PFKM):c.450+1G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Glycogen Storage Disease by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in at least one gene selected from GAA, AGL, PHKB, PRKAG2, G6PC, PGAM2, GBE1, PYGM, and PFKM, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00959] Cystic Fibrosis

[00960] In some embodiments, the methods, systems, and compositions described herein are used to correct one or more pathogenic G-to-A or C-to-T mutations/SNPs associated with Cystic Fibrosis. In some embodiment, the pathogenic mutations/SNPs are present in the CFTR gene, including at least the followings:

NM_000492.3(CFTR):c.3712C>T (p.Gln1238Ter)

NM_000492.3(CFTR):c.3484C>T (p.Arg1162Ter)

NM_000492.3(CFTR):c.1766+1G>A

NM_000492.3(CFTR):c.1477C>T (p.Gln493Ter)

NM_000492.3(CFTR):c.2538G>A (p.Trp846Ter)

NM_000492.3(CFTR):c.2551C>T (p.Arg851Ter)

NM_000492.3(CFTR):c.3472C>T (p.Arg1158Ter)

NM_000492.3(CFTR):c.1475C>T (p.Ser492Phe)

NM_000492.3(CFTR):c.1679G>A (p.Arg560Lys)

NM_000492.3(CFTR):c.3197G>A (p.Arg1066His)

NM_000492.3(CFTR):c.3873+1G>A

NM_000492.3(CFTR):c.3196C>T (p.Arg1066Cys)

NM_000492.3(CFTR):c.2490+1G>A

NM_000492.3(CFTR):c.3718-1G>A

NM_000492.3(CFTR):c.171G>A (p.Trp57Ter)

NM_000492.3(CFTR):c.3937C>T (p.Gln1313Ter)

NM_000492.3(CFTR):c.274G>A (p.Glu92Lys)

NM_000492.3(CFTR):c.1013C>T (p.Thr338Ile)

NM_000492.3(CFTR):c.3266G>A (p.Trp1089Ter)

NM_000492.3(CFTR):c.1055G>A (p.Arg352Gln)

NM_000492.3(CFTR):c.1654C>T (p.Gln552Ter)

NM_000492.3(CFTR):c.2668C>T (p.Gln890Ter)

NM_000492.3(CFTR):c.3611G>A (p.Trp1204Ter)

NM_000492.3(CFTR):c.1585-8G>A

NM_000492.3(CFTR):c.223C>T (p.Arg75Ter)

NM_000492.3(CFTR):c.1680-1G>A

NM_000492.3(CFTR):c.349C>T (p.Arg117Cys)

NM_000492.3(CFTR):c.1203G>A (p.Trp401Ter)

NM_000492.3(CFTR):c.1240C>T (p.Gln414Ter)

NM_000492.3(CFTR):c.1202G>A (p.Trp401Ter)

NM_000492.3(CFTR):c.1209+1G>A

NM_000492.3(CFTR):c.115C>T (p.Gln39Ter)

NM_000492.3(CFTR):c.1116+1G>A

NM_000492.3(CFTR):c.1393-1G>A

NM_000492.3(CFTR):c.1573C>T (p.Gln525Ter)

NM_000492.3(CFTR):c.164+1G>A

NM_000492.3(CFTR):c.166G>A (p.Glu56Lys)

NM_000492.3(CFTR):c.170G>A (p.Trp57Ter)

NM_000492.3(CFTR):c.2053C>T (p.Gln685Ter)

NM_000492.3(CFTR):c.2125C>T (p.Arg709Ter)

NM_000492.3(CFTR):c.2290C>T (p.Arg764Ter)

NM_000492.3(CFTR):c.2353C>T (p.Arg785Ter)

NM_000492.3(CFTR):c.2374C>T (p.Arg792Ter)

NM_000492.3(CFTR):c.2537G>A (p.Trp846Ter)

NM_000492.3(CFTR):c.292C>T (p.Gln98Ter)

NM_000492.3(CFTR):c.2989-1G>A

NM_000492.3(CFTR):c.3293G>A (p.Trp1098Ter)
NM_000492.3(CFTR):c.4144C>T (p.Gln1382Ter)
NM_000492.3(CFTR):c.4231C>T (p.Gln1411Ter)
NM_000492.3(CFTR):c.4234C>T (p.Gln1412Ter)
NM_000492.3(CFTR):c.579+5G>A
NM_000492.3(CFTR):c.595C>T (p.His199Tyr)
NM_000492.3(CFTR):c.613C>T (p.Pro205Ser)
NM_000492.3(CFTR):c.658C>T (p.Gln220Ter)
NM_000492.3(CFTR):c.1117-1G>A
NM_000492.3(CFTR):c.3294G>A (p.Trp1098Ter)
NM_000492.3(CFTR):c.1865G>A (p.Gly622Asp)
NM_000492.3(CFTR):c.743+1G>A
NM_000492.3(CFTR):c.1679+1G>A
NM_000492.3(CFTR):c.1657C>T (p.Arg553Ter)
NM_000492.3(CFTR):c.1675G>A (p.Ala559Thr)
NM_000492.3(CFTR):c.165-1G>A
NM_000492.3(CFTR):c.200C>T (p.Pro67Leu)
NM_000492.3(CFTR):c.2834C>T (p.Ser945Leu)
NM_000492.3(CFTR):c.3846G>A (p.Trp1282Ter)
NM_000492.3(CFTR):c.1652G>A (p.Gly551Asp)
NM_000492.3(CFTR):c.4426C>T (p.Gln1476Ter)
NM_000492.3:c.3718-2477C>T
NM_000492.3(CFTR):c.2988+1G>A
NM_000492.3(CFTR):c.2657+5G>A
NM_000492.3(CFTR):c.2988G>A (p.Gln996=)
NM_000492.3(CFTR):c.274-1G>A
NM_000492.3(CFTR):c.3612G>A (p.Trp1204Ter)
NM_000492.3(CFTR):c.1646G>A (p.Ser549Asn)
NM_000492.3(CFTR):c.3752G>A (p.Ser1251Asn)
NM_000492.3(CFTR):c.4046G>A (p.Gly1349Asp)
NM_000492.3(CFTR):c.532G>A (p.Gly178Arg)
NM_000492.3(CFTR):c.3731G>A (p.Gly1244Glu)
NM_000492.3(CFTR):c.1651G>A (p.Gly551Ser)
NM_000492.3(CFTR):c.1585-1G>A

NM_000492.3(CFTR):c.1000C>T (p.Arg334Trp)

NM_000492.3(CFTR):c.254G>A (p.Gly85Glu)

NM_000492.3(CFTR):c.1040G>A (p.Arg347His)

NM_000492.3(CFTR):c.273+1G>A

See Table A. Accordingly, an aspect of the invention relates to a method for treating or preventing Cystic Fibrosis by correcting one or more pathogenic G-to-A or C-to-T mutations/SNPs, particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs present in the CFTR gene, and more particularly one or more pathogenic G-to-A or C-to-T mutations/SNPs described above.

[00961] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 2 breast-ovarian cancer, wherein the pathogenic A>G mutation or SNP is located in the BRCA2 gene (HGVS: U43746.1:n.7829+1G>A). Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 2 breast-ovarian cancer by correcting the aforementioned pathogenic A>G mutation or SNP.

[00962] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with hereditary factor IX deficiency, wherein the pathogenic A>G mutation or SNP is located at GRCh38: ChrX: 139537145 in the F9 gene, which results in an Arg to Gln substitution. Accordingly, an additional aspect of the invention relates to a method for treating or preventing hereditary factor IX deficiency by correcting the aforementioned pathogenic A>G mutation or SNP.

[00963] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with beta-plus-thalassemia, beta thalassemia, and beta thalassemia major, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr11: 5226820 in the HBB gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing with beta-plus-thalassemia, beta thalassemia, and beta thalassemia major by correcting the aforementioned pathogenic A>G mutation or SNP.

[00964] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Marfan syndrome, wherein the pathogenic A>G mutation or SNP is located in the FBN1 gene (IVS2DS, G-A, +1), as reported by Yamamoto *et al.* J Hum Genet. 2000;45(2):115-8. Accordingly, an additional aspect of the invention relates to a method

for treating or preventing Marfan syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00965] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Wiskott-Aldrich syndrome, wherein the pathogenic A>G mutation or SNP is located at position -1 of intro 6 of the WAS gene (IVS6AS, G-A, -1), as reported by Kwan *et al.* (1995). Accordingly, an additional aspect of the invention relates to a method for treating or preventing Wiskott-Aldrich syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00966] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with cystic fibrosis, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr7:117590440 in the CFTR gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing cystic fibrosis by correcting the aforementioned pathogenic A>G mutation or SNP.

[00967] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with cystic fibrosis and hereditary pancreatitis, wherein the pathogenic A>G mutation or SNP is located GRCh38: Chr7:117606754 in the CFTR gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing cystic fibrosis and hereditary pancreatitis by correcting the aforementioned pathogenic A>G mutation or SNP.

[00968] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with cystic fibrosis, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr7: 117587738 in the CFTR gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing cystic fibrosis by correcting the aforementioned pathogenic A>G mutation or SNP.

[00969] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Turcot syndrome and Lynch syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr2:47470964 in the MSH2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing Turcot syndrome and Lynch syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00970] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with cystic fibrosis, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr7: 117642437 in the CFTR gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing cystic fibrosis by correcting the aforementioned pathogenic A>G mutation or SNP.

[00971] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome II and Lynch syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr3:37001058 in the MLH1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing Lynch syndrome II and Lynch syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00972] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with cystic fibrosis, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr7: 117642594 in the CFTR gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing cystic fibrosis by correcting the aforementioned pathogenic A>G mutation or SNP.

[00973] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with cystic fibrosis, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr7: 117592658 in the CFTR gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing cystic fibrosis by correcting the aforementioned pathogenic A>G mutation or SNP.

[00974] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr17:43057051 in the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00975] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with dihydropyrimidine dehydrogenase deficiency, Hirschsprung disease 1, fluorouracil response,
pyrimidine analogues response - toxicity/ADR, capecitabine response - toxicity/ADR, fluorouracil response - toxicity/ADR, tegafur response - toxicity/ADR, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr1:97450058 in the DPYD gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing dihydropyrimidine dehydrogenase deficiency, Hirschsprung disease 1, fluorouracil response,
pyrimidine analogues response - toxicity/ADR, capecitabine response - toxicity/ADR, fluorouracil response - toxicity/ADR, tegafur response - toxicity/ADR by correcting the aforementioned pathogenic A>G mutation or SNP.

[00976] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr2:47478520 in the MSH2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the Lynch syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00977] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr3:37011819 in the MLH1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the Lynch syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00978] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr3: 37014545 in the MLH1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the Lynch syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00979] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38:

Chr3: 37011867 in the MLH1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the Lynch syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00980] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr3: 37025636 in the MLH1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the Lynch syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00981] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr3: 37004475 in the MLH1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the Lynch syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00982] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome and hereditary cancer-predisposing syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr2:47416430 in the MSH2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing Lynch syndrome and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00983] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome and hereditary cancer-predisposing syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr2: 47408400 in the MSH2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing Lynch syndrome and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00984] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with Lynch syndrome and hereditary cancer-predisposing syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr3:36996710 in the MLH1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or

preventing Lynch syndrome and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00985] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 1 breast-ovarian cancer, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr17:43067696 in the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 1 breast-ovarian cancer by correcting the aforementioned pathogenic A>G mutation or SNP.

[00986] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 2 breast-ovarian cancer and hereditary breast and ovarian cancer syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr13:32356610 in the BRCA2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 2 breast-ovarian cancer and hereditary breast and ovarian cancer syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00987] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with primary dilated cardiomyopathy and primary familial hypertrophic cardiomyopathy, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr14:23419993 in the MYH7 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing primary dilated cardiomyopathy and primary familial hypertrophic cardiomyopathy by correcting the aforementioned pathogenic A>G mutation or SNP.

[00988] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with primary familial hypertrophic cardiomyopathy, camptocormism, and hypertrophic cardiomyopathy, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr14:23415225 in the MYH7 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing primary familial hypertrophic cardiomyopathy, camptocormism, and hypertrophic cardiomyopathy by correcting the aforementioned pathogenic A>G mutation or SNP.

[00989] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial cancer of breast, familial 2 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome, wherein the

pathogenic A>G mutation or SNP is located at GRCh38: Chr13:32357741 in the BRCA2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the familial cancer of breast, familial 2 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00990] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with primary dilated cardiomyopathy, hypertrophic cardiomyopathy, cardiomyopathy, and left ventricular noncompaction, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr14:23431584 in the MYH7 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing primary dilated cardiomyopathy, hypertrophic cardiomyopathy, cardiomyopathy, and left ventricular noncompaction by correcting the aforementioned pathogenic A>G mutation or SNP.

[00991] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr17:43067607 in the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00992] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, hereditary cancer-predisposing syndrome, and breast cancer, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr17:43047666 in the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, hereditary cancer-predisposing syndrome, and breast cancer by correcting the aforementioned pathogenic A>G mutation or SNP.

[00993] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 2 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome, wherein the pathogenic A>G mutation or SNP is

located at GRCh38: Chr13:32370558 in the BRCA2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 2 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00994] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, hereditary cancer-predisposing syndrome, and breast cancer, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr17:43074330 in the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, hereditary cancer-predisposing syndrome, and breast cancer by correcting the aforementioned pathogenic A>G mutation or SNP.

[00995] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic A-to-G (A>G) mutation or SNP believed to be associated with familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome, wherein the pathogenic A>G mutation or SNP is located at GRCh38: Chr17: 43082403 in the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 1 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic A>G mutation or SNP.

[00996] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic C-to-T (C>T) mutation or SNP believed to be associated with cystic fibrosis and hereditary pancreatitis, wherein the pathogenic C>T mutation or SNP is located at GRCh38: Chr7:117639961 in the CFTR gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the cystic fibrosis and hereditary pancreatitis by correcting the aforementioned pathogenic C>T mutation or SNP.

[00997] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic C-to-T (C>T) mutation or SNP believed to be associated with familial 2 breast-ovarian cancer, wherein the pathogenic C>T mutation or SNP is located at GRCh38: Chr13:32336492 in the BRCA2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the familial 2 breast-ovarian cancer by correcting the aforementioned pathogenic C>T mutation or SNP.

[00998] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic C-to-T (C>T) mutation or SNP believed to be associated with familial 1 breast-ovarian cancer, wherein the pathogenic C>T mutation or SNP is located at GRCh38: Chr17:43063365 in the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the familial 1 breast-ovarian cancer by correcting the aforementioned pathogenic C>T mutation or SNP.

[00999] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic C-to-T (C>T) mutation or SNP believed to be associated with familial 1 breast-ovarian cancer, wherein the pathogenic C>T mutation or SNP is located at GRCh38: Chr17:43093613 in the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing the familial 1 breast-ovarian cancer by correcting the aforementioned pathogenic C>T mutation or SNP.

[001000] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic C-to-T (C>T) mutation or SNP believed to be associated with familial cancer of breast, and familial 1 breast-ovarian cancer, wherein the pathogenic C>T mutation or SNP is located at at GRCh38: Chr17:43093931 of the BRCA1 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial cancer of breast, and familial 1 breast-ovarian cancer by correcting the aforementioned pathogenic C>T mutation or SNP.

[001001] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic C-to-T (C>T) mutation or SNP believed to be associated with familial hypertrophic cardiomyopathy 1, primary familial hypertrophic cardiomyopathy, and hypertrophic cardiomyopathy, wherein the pathogenic C>T mutation or SNP is located at GRCh38: Chr14:23429279 of the MYH7 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial hypertrophic cardiomyopathy 1, primary familial hypertrophic cardiomyopathy, and hypertrophic cardiomyopathy by correcting the aforementioned pathogenic C>T mutation or SNP.

[001002] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic C-to-T (C>T) mutation or SNP believed to be associated with familial 2 breast-ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome, wherein the pathogenic C>T mutation or SNP is located at GRCh38: Chr13:32356472 of the BRCA2 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial 2 breast-

ovarian cancer, hereditary breast and ovarian cancer syndrome, and hereditary cancer-predisposing syndrome by correcting the aforementioned pathogenic C>T mutation or SNP.

[001003] In some embodiments, the methods, systems, and compositions described herein are used to correct a pathogenic C-to-T (C>T) mutation or SNP believed to be associated with familial hypertrophic cardiomyopathy 1, primary familial hypertrophic cardiomyopathy, familial restrictive cardiomyopathy, and hypertrophic cardiomyopathy, wherein the pathogenic C>T mutation or SNP is located at GRCh38: Chr14:23429005 in the MYH7 gene. Accordingly, an additional aspect of the invention relates to a method for treating or preventing familial hypertrophic cardiomyopathy 1, primary familial hypertrophic cardiomyopathy, familial restrictive cardiomyopathy, and hypertrophic cardiomyopathy by correcting the aforementioned pathogenic C>T mutation or SNP.

[001004] Additional pathogenic A>G mutations and SNPs are found in the ClinVar database and listed in Table A. Accordingly, an additional aspect of the present disclosure relates to correction of a pathogenic A>G mutation or SNP listed in Table A using the methods, systems, and compositions described herein to treat or prevent a disease or condition associated therewith.

[001005] Additional pathogenic C>T mutations and SNPs are found in the ClinVar database and also listed in Table A. Accordingly, an additional aspect of the present disclosure relates to correction of a pathogenic C>T mutation or SNP listed in Table A using the methods, systems, and compositions described herein to treat or prevent a disease or condition associated therewith.

TABLE A - Pathogenic G-to-A and C-to T Mutations/SNPs

Name	GeneSymbol	Chr	Location	Disease-Associated Phenotype
NM_000197.1(HSD17B3):c.238C>T (p.Arg80Trp)	HSD17B3	9	96254907	17-Beta-Hydroxysteroid Dehydrogenase III Deficiency;Testosterone 17-beta-dehydrogenase deficiency
NM_001287341.1(NADK2):c.595C>T (p.Arg199Ter)	NADK2	5	36200275	2,4-Dienoyl-CoA reductase deficiency
NM_000500.7(CYP21A2):c.1069C>T (p.Arg357Trp)	CYP21A2	6	32040535	21-hydroxylase deficiency
NM_000500.7(CYP21A2):c.92C>T (p.Pro31Leu)	CYP21A2	6	32038514	21-hydroxylase deficiency
NM_000500.7(CYP21A2):c.874G>A (p.Gly292Ser)	CYP21A2	6	32040140	21-hydroxylase deficiency
NM_000500.7(CYP21A2):c.1360C>T (p.Pro454Ser)	CYP21A2	6	32041006	21-hydroxylase deficiency
NM_000500.7(CYP21A2):c.955C>T (p.Gln319Ter)	CYP21A2	6	32040421	21-hydroxylase deficiency
NM_000500.7(CYP21A2):c.1217G>A (p.Trp406Ter)	CYP21A2	6	32040766	21-hydroxylase deficiency
NM_018706.6(DHTKD1):c.1228C>T (p.Arg410Ter)	DHTKD1	10	12094141	2-aminoadipic 2-oxoadipic aciduria
NM_018706.6(DHTKD1):c.2185G>A (p.Gly729Arg)	DHTKD1	10	12112930	2-aminoadipic 2-oxoadipic aciduria;not provided
NM_001037811.2(HSD17B10):c.388C>T (p.Arg130Cys)	HSD17B10	X	53432086	2-methyl-3-hydroxybutyric aciduria
NM_001166120.1(HSD3B2):c.512G>A (p.Trp171Ter)	HSD3B2	1	119422013	3 beta-Hydroxysteroid dehydrogenase deficiency
NM_001166120.1(HSD3B2):c.424G>A (p.Glu142Lys)	HSD3B2	1	119421925	3 beta-Hydroxysteroid dehydrogenase deficiency
NM_001166120.1(HSD3B2):c.776C>T (p.Thr259Met)	HSD3B2	1	119422277	3 beta-Hydroxysteroid dehydrogenase deficiency
NM_001166120.1(HSD3B2):c.1022C>T (p.Pro341Leu)	HSD3B2	1	119422523	3 beta-Hydroxysteroid dehydrogenase deficiency
NM_020166.4(MCCC1):c.1604C>T (p.Ser535Phe)	MCCC1	3	183034068	3 Methylcrotonyl-CoA carboxylase 1 deficiency
NM_020166.4(MCCC1):c.1114C>T (p.Gln372Ter)	MCCC1	3	183041720	3 Methylcrotonyl-CoA carboxylase 1 deficiency
NM_020166.4(MCCC1):c.841C>T (p.Arg281Ter)	MCCC1	3	183057343	3 Methylcrotonyl-CoA carboxylase 1 deficiency
NM_020166.4(MCCC1):c.640-1G>A	MCCC1	3	183071121	3 Methylcrotonyl-CoA carboxylase 1 deficiency;not provided
NM_022132.4(MCCC2):c.464G>A (p.Arg155Gln)	MCCC2	5	71602586	3-methylcrotonyl CoA carboxylase 2 deficiency
NM_022132.4(MCCC2):c.1574+1G>A	MCCC2	5	71652755	3-methylcrotonyl CoA carboxylase 2 deficiency
NM_022132.4(MCCC2):c.1081C>T (p.Arg361Ter)	MCCC2	5	71643827	3-methylcrotonyl CoA carboxylase 2 deficiency
NM_022132.4(MCCC2):c.214C>T (p.Arg72Ter)	MCCC2	5	71596297	3-methylcrotonyl CoA carboxylase 2 deficiency
NM_022132.4(MCCC2):c.994C>T (p.Arg332Ter)	MCCC2	5	71635241	3-methylcrotonyl CoA carboxylase 2 deficiency;not provided
NM_022132.4(MCCC2):c.568C>T (p.His190Tyr)	MCCC2	5	71604412	3-methylcrotonyl CoA carboxylase 2 deficiency;not provided
NM_022132.4(MCCC2):c.1015G>A (p.Val339Met)	MCCC2	5	71641018	3-methylcrotonyl CoA carboxylase 2 deficiency;not provided
NM_001698.2(AUH):c.589C>T (p.Arg197Ter)	AUH	9	91297993	3-Methylglutaconic aciduria
NM_001698.2(AUH):c.895-1G>A	AUH	9	91216107	3-Methylglutaconic aciduria
NM_001698.2(AUH):c.650G>A (p.Gly217Asp)	AUH	9	91296026	3-Methylglutaconic aciduria
NM_000116.4(TAZ):c.589G>A (p.Gly197Arg)	TAZ	X	154420037	3-Methylglutaconic aciduria type 2
NM_000116.4(TAZ):c.718G>A (p.Gly240Arg)	TAZ	X	154420676	3-Methylglutaconic aciduria type 2;not provided
NM_025136.3(OPA3):c.415C>T (p.Gln139Ter)	OPA3	19	45553639	3-Methylglutaconic aciduria type 3
NM_030813.5(CLPB):c.1772C>T (p.Ala591Val)	CLPB	11	72294125	3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia
NM_030813.5(CLPB):c.1233G>A (p.Met411Ile)	CLPB	11	72302328	3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia
NM_030813.5(CLPB):c.1249C>T (p.Arg417Ter)	CLPB	11	72302312	3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia
NM_030813.5(CLPB):c.803C>T (p.Thr268Met)	CLPB	11	72358942	3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia
NM_030813.5(CLPB):c.1915G>A (p.Glu639Lys)	CLPB	11	72293576	3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia
NM_030813.5(CLPB):c.1501G>A (p.Glu501Lys)	CLPB	11	72295567	3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia
NM_030813.5(CLPB):c.805G>A (p.Ala269Thr)	CLPB	11	72358940	3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia
NM_030813.5(CLPB):c.748C>T (p.Arg250Ter)	CLPB	11	72358997	3-methylglutaconic aciduria with cataracts, neurologic involvement, and neutropenia
NM_032861.3(SERAC1):c.202C>T (p.Arg68Ter)	SERAC1	6	158150516	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome
NM_032861.3(SERAC1):c.442C>T (p.Arg148Ter)	SERAC1	6	158146827	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome;not provided

NM_013247.4(HTRA2):c.1211G>A (p.Arg404Gln)	HTRA2	2	74532714	3-METHYLGLUTACONIC ACIDURIA, TYPE VIII
NM_001698.2(AUH):c.559G>A (p.Gly187Ser)	AUH	9	91298023	3-Methylglutaconic aciduria;not provided
NM_000348.3(SRD5A2):c.736C>T (p.Arg246Trp)	SRD5A2	2	31526225	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency
NM_000348.3(SRD5A2):c.344G>A (p.Gly115Asp)	SRD5A2	2	31533704	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency
NM_000348.3(SRD5A2):c.547G>A (p.Gly183Ser)	SRD5A2	2	31531371	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency
NM_000348.3(SRD5A2):c.679C>T (p.Arg227Ter)	SRD5A2	2	31529326	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency
NM_000348.3(SRD5A2):c.682G>A (p.Ala228Thr)	SRD5A2	2	31529323	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency
NM_000348.3(SRD5A2):c.586G>A (p.Gly196Ser)	SRD5A2	2	31529419	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency
NM_000348.3(SRD5A2):c.211C>T (p.Gln71Ter)	SRD5A2	2	31580690	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency
NM_000348.3(SRD5A2):c.680G>A (p.Arg227Gln)	SRD5A2	2	31529325	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency;Micropenis;not provided
NM_012082.3(ZFPM2):c.779G>A (p.Arg260Gln)	ZFPM2	8	105798763	46,XY sex reversal 9
NM_003140.2(SRY):c.277C>T (p.Gln93Ter)	SRY	Y	2787327	46,XY sex reversal, type 1
NM_003140.2(SRY):c.337G>A (p.Ala113Thr)	SRY	Y	2787267	46,XY sex reversal, type 1
NM_003140.2(SRY):c.320G>A (p.Trp107Ter)	SRY	Y	2787284	46,XY sex reversal, type 1
NM_003140.2(SRY):c.192G>A (p.Met64Ile)	SRY	Y	2787412	46,XY sex reversal, type 1
NM_003140.2(SRY):c.209G>A (p.Trp70Ter)	SRY	Y	2787395	46,XY sex reversal, type 1
NM_003140.2(SRY):c.397C>T (p.Arg133Trp)	SRY	Y	2787207	46,XY sex reversal, type 1
NM_003140.2(SRY):c.4C>T (p.Gln2Ter)	SRY	Y	2787600	46,XY sex reversal, type 1
NM_003140.2(SRY):c.53G>A (p.Ser18Asn)	SRY	Y	2787551	46,XY sex reversal, type 1
NM_003140.2(SRY):c.284G>A (p.Gly95Glu)	SRY	Y	2787320	46,XY sex reversal, type 1
NM_004959.4(NR5A1):c.275G>A (p.Arg92Gln)	NR5A1	9	124500685	46,XY sex reversal, type 3
NM_004959.4(NR5A1):c.43G>A (p.Val15Met)	NR5A1	9	124503353	46,XY sex reversal, type 3
NM_004959.4(NR5A1):c.234G>A (p.Met78Ile)	NR5A1	9	124503089	46,XY sex reversal, type 3
NM_004959.4(NR5A1):c.271G>A (p.Gly91Ser)	NR5A1	9	124500689	46,XY sex reversal, type 3
NM_004959.4(NR5A1):c.877G>A (p.Asp293Asn)	NR5A1	9	124493143	46,XY sex reversal, type 3;Premature ovarian failure 7
NM_004959.4(NR5A1):c.3G>A (p.Met1Ile)	NR5A1	9	124503393	46,XY sex reversal, type 3;Premature ovarian failure 7
NM_005189.2(CBX2):c.293C>T (p.Pro98Leu)	CBX2	17	79783736	46,XY sex reversal, type 5
NM_017570.4(OPLAH):c.3265G>A (p.Val1089Ile)	OPLAH	8	144052487	5-Oxoprolinase deficiency
NM_000317.2(PTS):c.74G>A (p.Arg25Gln)	PTS	11	112226517	6-pyruvoyl-tetrahydropterin synthase deficiency
NM_000317.2(PTS):c.259C>T (p.Pro87Ser)	PTS	11	112233178	6-pyruvoyl-tetrahydropterin synthase deficiency
NM_000317.2(PTS):c.166G>A (p.Val56Met)	PTS	11	112230210	6-pyruvoyl-tetrahydropterin synthase deficiency
NM_000317.2(PTS):c.286G>A (p.Asp96Asn)	PTS	11	112233305	6-pyruvoyl-tetrahydropterin synthase deficiency
NM_004463.2(FGD1):c.1565G>A (p.Arg522His)	FGD1	X	54465522	Aarskog syndrome
NM_004463.2(FGD1):c.1223G>A (p.Arg408Gln)	FGD1	X	54467901	Aarskog syndrome
NM_004463.2(FGD1):c.1966C>T (p.Arg656Ter)	FGD1	X	54455497	Aarskog syndrome
NM_004463.2(FGD1):c.1829G>A (p.Arg610Gln)	FGD1	X	54456233	Aarskog syndrome;not provided
NM_004463.2(FGD1):c.1468C>T (p.Gln490Ter)	FGD1	X	54465725	Aarskog syndrome;not provided
NM_004463.2(FGD1):c.175C>T (p.Gln59Ter)	FGD1	X	54495258	Aarskog syndrome;not provided
NM_000969.4(RPL5):c.67C>T (p.Arg23Ter)	RPL5	1	92833452	Aase syndrome
NM_000969.4(RPL5):c.418G>A (p.Gly140Ser)	RPL5	1	92836283	Aase syndrome
NM_000969.4(RPL5):c.664C>T (p.Gln222Ter)	RPL5	1	92837592	Aase syndrome
NM_000350.2(ABCA4):c.5882G>A (p.Gly1961Glu)	ABCA4	1	94008251	ABCA4-Related Disorders;Cone-rod dystrophy 3;Stargardt disease 1;not provided
NM_000115.4(EDNRB):c.601C>T (p.Arg201Ter)	EDNRB	13	77903356	ABCD syndrome
NM_000309.4(PPOX):c.1281G>A (p.Trp427Ter)	PPOX	1	161170939	Abdominal colic;Abnormal blistering of the skin;Porphyrinuria
NM_004714.2(DYRK1B):c.304C>T (p.Arg102Cys)	DYRK1B	19	39830443	Abdominal obesity-metabolic syndrome 3

NM_000253.3(MTTP):c.1783C>T (p.Arg595Ter)	MTTP	4	99611156	Abetalipoproteinaemia
NM_000253.3(MTTP):c.1619G>A (p.Arg540His)	MTTP	4	99608827	Abetalipoproteinaemia
NM_057179.2(TWIST2):c.223G>A (p.Glu75Lys)	TWIST2	2	238848438	Ablepharon macrostomia syndrome
NM_014874.3(MFN2):c.280C>T (p.Arg94Trp)	MFN2	1	11992659	Abnormal blistering of the skin;Abnormality of dental enamel;Abnormality of the teeth;Alopecia of scalp;Charcot-Marie-Tooth disease, type 2;Charcot-Marie-Tooth disease, type 2A2;Decreased body weight;Distal muscle weakness;EMG abnormality;Failure to thrive;Hereditary motor and sensory neuropathy with optic atrophy;Hyperpigmentation of the skin;Microcephaly;Nail dystrophy;Scarring;Scarring alopecia of scalp;Short stature;not provided
NM_000094.3(COL7A1):c.706C>T (p.Arg236Ter)	COL7A1	3	48592915	Abnormal blistering of the skin;Abnormality of dental enamel;Abnormality of the teeth;Alopecia of scalp;Decreased body weight;Distal muscle weakness;EMG abnormality;Epidermolysis bullosa dystrophica inversa, autosomal recessive;Failure to thrive;Hyperpigmentation of the skin;Microcephaly;Nail dystrophy;Recessive dystrophic epidermolysis bullosa;Scarring;Scarring alopecia of scalp;Short stature
NM_000094.3(COL7A1):c.6205C>T (p.Arg2069Cys)	COL7A1	3	48575218	Abnormal blistering of the skin;Abnormality of dental enamel;Abnormality of the teeth;Alopecia of scalp;Decreased body weight;Distal muscle weakness;EMG abnormality;Epidermolysis bullosa dystrophica inversa, autosomal recessive;Failure to thrive;Hyperpigmentation of the skin;Microcephaly;Nail dystrophy;Scarring;Scarring alopecia of scalp;Short stature
NM_000329.2(RPE65):c.1338+1G>A	RPE65	1	68431281	Abnormal electroretinogram;Abnormality of vision;Congenital blindness;Retinal degeneration
m.3700G>A	MT-ND1	MT	3700	Abnormal electroretinogram;Leber's optic atrophy;Optic neuropathy;Visual loss
NM_005120.2(MED12):c.2881C>T (p.Arg961Trp)	MED12	X	71127367	Abnormal facial shape;Agenesis of corpus callosum;Anal atresia;Broad thumb;FG syndrome;Global developmental delay;Intellectual disability
NM_139242.3(MTFMT):c.626C>T (p.Ser209Leu)	MTFMT	15	65021533	Abnormal facial shape;Combined oxidative phosphorylation deficiency 15;Decreased activity of cytochrome C oxidase in muscle tissue;Decreased activity of mitochondrial complex I;Inability to walk by childhood/adolescence;Leigh syndrome;Leigh syndrome due to mitochondrial complex I deficiency;Poor speech;Short stature;not provided
NM_018486.2(HDAC8):c.496C>T (p.Arg166Ter)	HDAC8	X	72495210	Abnormal facial shape;Delayed speech and language development;Global developmental delay;Intrauterine growth retardation;Microcephaly;Sparse scalp hair;not provided
NM_024757.4(EHMT1):c.2712+1G>A	EHMT1	9	137800985	Abnormal facial shape;Difficulty walking;Global developmental delay;Polymicrogyria
NM_003588.3(CUL4B):c.1162C>T (p.Arg388Ter)	CUL4B	X	120544179	Abnormal facial shape;Global developmental delay;Intellectual disability;Seizures;Short stature;Syndromic X-linked mental retardation, Cabezas type
NM_003742.2(ABCB11):c.3148C>T (p.Arg1050Cys)	ABCB11	2	168932442	Abnormal liver function tests during pregnancy;Intrahepatic cholestasis;Pruritus
NM_052867.3(NALCN):c.3390G>A (p.Pro1130=)	NALCN	13	101089846	Abnormal pattern of respiration;Cachexia;Intellectual disability, severe;Seizures;Strabismus
NM_014363.5(SACS):c.2182C>T (p.Arg728Ter)	SACS	13	23353788	Abnormality of brain morphology;not provided
NM_001360.2(DHCR7):c.278C>T (p.Thr93Met)	DHCR7	11	71444036	Abnormality of brain morphology;Smith-Lemli-Opitz syndrome;not provided
NM_005257.5(GATA6):c.1366C>T (p.Arg456Cys)	GATA6	18	22181516	Abnormality of cardiovascular system morphology;Congenital diaphragmatic hernia;Pancreatic agenesis and congenital heart disease
NM_152296.4(ATP1A3):c.410C>T (p.Ser137Phe)	ATP1A3	19	41986177	Abnormality of earlobe;Alternating hemiplegia of childhood 2;Depressed nasal bridge;Epicanthus;Seizures;Ventriculomegaly
NM_001376.4(DYNC1H1):c.1738G>A (p.Glu580Lys)	DYNC1H1	14	101985963	Abnormality of neuronal migration
NM_133436.3(ASNS):c.1648C>T (p.Arg550Cys)	ASNS	7	97852297	Abnormality of neuronal migration;Asparagine synthetase deficiency
NM_006009.3(TUBA1A):c.1148C>T (p.Ala383Val)	TUBA1A	12	49185218	Abnormality of neuronal migration;Lissencephaly 3
NM_007313.2(ABL1):c.1066G>A (p.Ala356Thr)	ABL1	9	130872961	Abnormality of skeletal morphology;Congenital heart disease;Failure to thrive

NM_015338.5(ASXL1):c.1210C>T (p.Arg404Ter)	ASXL1	20	32433408	Abnormality of the corpus callosum;C-like syndrome;Delayed gross motor development;Delayed speech and language development;Developmental delay;Feeding difficulties;Glabellar hemangioma;Global developmental delay;Hypertrichosis;Intellectual disability, severe;Prominent metopic ridge;Small for gestational age;dystrophia
NM_000462.3(UBE3A):c.2281G>A (p.Gly761Arg)	UBE3A	15	25354536	Abnormality of the corpus callosum;EEG abnormality;Expressive language delay;Global developmental delay;Poor speech;Seizures
NM_001282531.2(ADNP):c.2188C>T (p.Arg730Ter)	ADNP	20	50892526	Abnormality of the teeth;Agenesis of corpus callosum;Aggressive behavior;Global developmental delay;Growth hormone deficiency;Helsmoortel-van der aa syndrome;Hypothyroidism;Seizures;Stereotypy
NM_130837.2(OPA1):c.2635C>T (p.Arg879Ter)	OPA1	3	193662936	Abortive cerebellar ataxia
NM_130837.2(OPA1):c.1369G>A (p.Val457Met)	OPA1	3	193643436	Abortive cerebellar ataxia
NM_020964.2(EPG5):c.4588C>T (p.Gln1530Ter)	EPG5	18	45901054	Absent corpus callosum cataract immunodeficiency
NM_020964.2(EPG5):c.3481C>T (p.Arg1161Ter)	EPG5	18	45916110	Absent corpus callosum cataract immunodeficiency
NM_020964.2(EPG5):c.6232C>T (p.Arg2078Ter)	EPG5	18	45867742	Absent corpus callosum cataract immunodeficiency
NM_020964.2(EPG5):c.7447C>T (p.Arg2483Ter)	EPG5	18	45855683	Absent corpus callosum cataract immunodeficiency
NM_001396.3(DYRK1A):c.613C>T (p.Arg205Ter)	DYRK1A	21	37486563	Absent or delayed speech development;Deeply set eye;Feeding difficulties;Intellectual disability;Mental retardation, autosomal dominant 7;Microcephaly
NM_001396.3(DYRK1A):c.1309C>T (p.Arg437Ter)	DYRK1A	21	37505352	Absent or delayed speech development;Deeply set eye;Feeding difficulties;Intellectual disability;Mental retardation, autosomal dominant 7;Microcephaly;Seizures;not provided
NM_001396.3(DYRK1A):c.763C>T (p.Arg255Ter)	DYRK1A	21	37490273	Absent or delayed speech development;Deeply set eye;Feeding difficulties;Intellectual disability;Microcephaly;Seizures
NM_020732.3(ARID1B):c.2941C>T (p.Gln981Ter)	ARID1B	6	157167101	Absent speech;Blepharophimosis;Intellectual disability, moderate;Long eyelashes;Thick lower lip vermilion;Thin upper lip vermilion
NM_007075.3(WDR45):c.700C>T (p.Arg234Ter)	WDR45	X	49075573	Absent speech;Delayed gross motor development;Delayed speech and language development;Global developmental delay;Hypoplasia of the corpus callosum;Neurodegeneration with brain iron accumulation 5;Seizures;not provided
NM_000346.3(SOX9):c.493C>T (p.His165Tyr)	SOX9	17	72122780	Acampomelic campomelic dysplasia
NM_000342.3(SLC4A1):c.2603C>T (p.Pro868Leu)	SLC4A1	17	44251211	Acanthocytosis due to band 3 ht
NM_015665.5(AAAS):c.1288C>T (p.Leu430Phe)	AAAS	12	53308095	Achalasia-alacrima syndrome
NM_004239.4(TRIP11):c.790C>T (p.Arg264Ter)	TRIP11	14	92015729	Achondrogenesis, type IA
NM_000112.3(SLC26A2):c.532C>T (p.Arg178Ter)	SLC26A2	5	149978184	Achondrogenesis, type IB;Atelosteogenesis type 2;Diastrophic dysplasia;Multiple epiphyseal dysplasia 4;not provided
NM_001844.4(COL2A1):c.2905G>A (p.Gly969Ser)	COL2A1	12	47978389	Achondrogenesis, type II
NM_001844.4(COL2A1):c.1547G>A (p.Gly516Asp)	COL2A1	12	47985946	Achondrogenesis, type II
NM_000142.4(FGFR3):c.1138G>A (p.Gly380Arg)	FGFR3	4	1804392	Achondroplasia;Epidermal nevus;not provided
NM_001298.2(CNGA3):c.488C>T (p.Pro163Leu)	CNGA3	2	98389696	Achromatopsia 2
NM_001298.2(CNGA3):c.848G>A (p.Arg283Gln)	CNGA3	2	98396018	Achromatopsia 2
NM_001298.2(CNGA3):c.1228C>T (p.Arg410Trp)	CNGA3	2	98396398	Achromatopsia 2
NM_001298.2(CNGA3):c.829C>T (p.Arg277Cys)	CNGA3	2	98395999	Achromatopsia 2
NM_001298.2(CNGA3):c.1306C>T (p.Arg436Trp)	CNGA3	2	98396476	Achromatopsia 2
NM_001298.2(CNGA3):c.847C>T (p.Arg283Trp)	CNGA3	2	98396017	Achromatopsia 2;Monochromacy
NM_001298.2(CNGA3):c.1585G>A (p.Val529Met)	CNGA3	2	98396755	Achromatopsia 2;not provided
NM_001298.2(CNGA3):c.101+1G>A	CNGA3	2	98370077	Achromatopsia 2;not provided
NM_019098.4(CNGB3):c.1304C>T (p.Ser435Phe)	CNGB3	8	86632768	Achromatopsia 3
NM_019098.4(CNGB3):c.607C>T (p.Arg203Ter)	CNGB3	8	86668055	Achromatopsia 3

NM_019098.4(CNGB3):c.1578+1G>A	CNGB3	8	86625982	Achromatopsia 3
NM_019098.4(CNGB3):c.1119G>A (p.Trp373Ter)	CNGB3	8	86643810	Achromatopsia 3;not provided
NM_005272.3(GNAT2):c.235C>T (p.Gln79Ter)	GNAT2	1	109610108	Achromatopsia 4
NM_005272.3(GNAT2):c.461+24G>A	GNAT2	1	109608607	Achromatopsia 4
NM_006204.3(PDE6C):c.2368G>A (p.Glu790Lys)	PDE6C	10	93663028	Achromatopsia 5
NM_006204.3(PDE6C):c.826C>T (p.Arg276Ter)	PDE6C	10	93622034	Achromatopsia 5
NM_007348.3(ATF6):c.970C>T (p.Arg324Cys)	ATF6	1	161819693	Achromatopsia 7
NM_000489.4(ATRX):c.20+1G>A	ATRX	X	77785981	Acquired hemoglobin H disease
NM_032737.3(LMNB2):c.1279G>A (p.Ala427Thr)	LMNB2	19	2434029	Acquired partial lipodystrophy;not provided
NM_032737.3(LMNB2):c.265-6C>T	LMNB2	19	2444546	Acquired partial lipodystrophy;not provided
NM_000218.2(KCNQ1):c.1747C>T (p.Arg583Cys)	KCNQ1	11	2777990	Acquired susceptibility to long QT syndrome 1;Congenital long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype;not provided
NM_198525.2(KIF7):c.460C>T (p.Arg154Ter)	KIF7	15	89649810	Acrocallosal syndrome, Schinzel type
NM_198525.2(KIF7):c.3001C>T (p.Gln1001Ter)	KIF7	15	89631605	Acrocallosal syndrome, Schinzel type
NM_198525.2(KIF7):c.3331C>T (p.Arg1111Ter)	KIF7	15	89629561	Acrocallosal syndrome, Schinzel type
NM_198525.2(KIF7):c.2917C>T (p.Arg973Ter)	KIF7	15	89631689	Acrocallosal syndrome, Schinzel type
NM_198525.2(KIF7):c.61C>T (p.Arg21Ter)	KIF7	15	89652870	Acrocallosal syndrome, Schinzel type;not provided
NM_002181.3(IHH):c.137C>T (p.Pro46Leu)	IHH	2	219060331	Acrocapitofemoral dysplasia
NM_002734.4(PRKAR1A):c.1102C>T (p.Arg368Ter)	PRKAR1A	17	68530405	Acrodysostosis 1 with or without hormone resistance
NM_001165899.1(PDE4D):c.1835G>A (p.Gly612Asp)	PDE4D	5	58975076	Acrodysostosis 2, with or without hormone resistance
NM_001165899.1(PDE4D):c.728C>T (p.Ala243Val)	PDE4D	5	59038869	Acrodysostosis 2, with or without hormone resistance
NM_020427.2(SLURP1):c.178+1G>A	SLURP1	8	142741802	Acroerythrokeratoderma
NM_020427.2(SLURP1):c.286C>T (p.Arg96Ter)	SLURP1	8	142741169	Acroerythrokeratoderma
NM_020427.2(SLURP1):c.256G>A (p.Gly86Arg)	SLURP1	8	142741199	Acroerythrokeratoderma
NM_020427.2(SLURP1):c.296G>A (p.Cys99Tyr)	SLURP1	8	142741159	Acroerythrokeratoderma
NM_001681.3(ATP2A2):c.1805C>T (p.Pro602Leu)	ATP2A2	12	110340702	Acrokeratosis verruciformis of Hopf
NM_001681.3(ATP2A2):c.2093C>T (p.Ala698Val)	ATP2A2	12	110340990	Acrokeratosis verruciformis of Hopf
NM_020928.1(ZSWIM6):c.3487C>T (p.Arg1163Trp)	ZSWIM6	5	61544156	Acromelic frontonasal dysostosis;not provided
NM_003995.3(NPR2):c.1162C>T (p.Arg388Ter)	NPR2	9	35800427	Acromesomelic dysplasia Maroteaux type
NM_003995.3(NPR2):c.1435C>T (p.Arg479Ter)	NPR2	9	35801153	Acromesomelic dysplasia Maroteaux type
NM_003995.3(NPR2):c.2944G>A (p.Asp982Asn)	NPR2	9	35808811	Acromesomelic dysplasia Maroteaux type
NM_000138.4(FBN1):c.5182G>A (p.Ala1728Thr)	FBN1	15	48463124	Acromicric dysplasia;Geleophysic dysplasia 2
NM_005149.2(TBX19):c.856C>T (p.Arg286Ter)	TBX19	1	168305136	ACTH deficiency
NM_005149.2(TBX19):c.383C>T (p.Ser128Phe)	TBX19	1	168291339	ACTH deficiency
NM_000529.2(MC2R):c.601C>T (p.Arg201Ter)	MC2R	18	13884918	ACTH resistance
NM_000529.2(MC2R):c.382C>T (p.Arg128Cys)	MC2R	18	13885137	ACTH resistance
NM_000529.2(MC2R):c.319G>A (p.Asp107Asn)	MC2R	18	13885200	ACTH resistance
NM_000529.2(MC2R):c.409C>T (p.Arg137Trp)	MC2R	18	13885110	ACTH resistance
NM_001288767.1(ARMC5):c.1084C>T (p.Arg362Ter)	ARMC5	16	31462346	Acth-independent macronodular adrenal hyperplasia 2
NM_001288767.1(ARMC5):c.2977C>T (p.Arg993Trp)	ARMC5	16	31466773	Acth-independent macronodular adrenal hyperplasia 2
NM_001288767.1(ARMC5):c.541C>T (p.Gln181Ter)	ARMC5	16	31459780	Acth-independent macronodular adrenal hyperplasia 2
NM_001288767.1(ARMC5):c.2062C>T (p.Arg688Trp)	ARMC5	16	31464800	Acth-independent macronodular adrenal hyperplasia 2
NM_000690.3(ALDH2):c.1510G>A (p.Glu504Lys)	ALDH2	12	111803962	Acute alcohol sensitivity;Alcohol dependence;Esophageal cancer, alcohol-related, susceptibility to;Sublingual nitroglycerin, susceptibility to poor response to;Susceptibility to hangover
NM_000190.3(HMBS):c.77G>A (p.Arg26His)	HMBS	11	119088298	Acute intermittent porphyria

NM_000190.3(HMBS):c.346C>T (p.Arg116Trp)	HMBS	11	119089991	Acute intermittent porphyria
NM_000190.3(HMBS):c.518G>A (p.Arg173Gln)	HMBS	11	119091432	Acute intermittent porphyria
NM_000190.3(HMBS):c.463C>T (p.Gln155Ter)	HMBS	11	119090230	Acute intermittent porphyria
NM_000190.3(HMBS):c.446G>A (p.Arg149Gln)	HMBS	11	119090213	Acute intermittent porphyria
NM_000190.3(HMBS):c.593G>A (p.Trp198Ter)	HMBS	11	119091507	Acute intermittent porphyria
NM_000190.3(HMBS):c.91G>A (p.Ala31Thr)	HMBS	11	119088638	Acute intermittent porphyria
NM_000190.3(HMBS):c.499C>T (p.Arg167Trp)	HMBS	11	119091413	Acute intermittent porphyria
NM_000190.3(HMBS):c.331G>A (p.Gly111Arg)	HMBS	11	119089747	Acute intermittent porphyria
NM_000190.3(HMBS):c.667G>A (p.Glu223Lys)	HMBS	11	119092419	Acute intermittent porphyria
NM_000190.3(HMBS):c.748G>A (p.Glu250Lys)	HMBS	11	119092500	Acute intermittent porphyria
NM_000190.3(HMBS):c.754G>A (p.Ala252Thr)	HMBS	11	119092506	Acute intermittent porphyria
NM_000190.3(HMBS):c.755C>T (p.Ala252Val)	HMBS	11	119092507	Acute intermittent porphyria
NM_000190.3(HMBS):c.647G>A (p.Gly216Asp)	HMBS	11	119092159	Acute intermittent porphyria
NM_000190.3(HMBS):c.445C>T (p.Arg149Ter)	HMBS	11	119090212	Acute intermittent porphyria
NM_000190.3(HMBS):c.601C>T (p.Arg201Trp)	HMBS	11	119091515	Acute intermittent porphyria;not provided
NM_000190.3(HMBS):c.849G>A (p.Trp283Ter)	HMBS	11	119092958	Acute intermittent porphyria;not provided
NM_004119.2(FLT3):c.2503G>A (p.Asp835Asn)	FLT3	13	28018505	Acute myeloid leukemia
NM_000546.5(TP53):c.818G>A (p.Arg273His)	TP53	17	7673802	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Adrenocortical Carcinoma;Brainstem glioma;Chronic lymphocytic leukemia;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni syndrome 1;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Medulloblastoma;Multiple myeloma;Neoplasm;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Small cell lung cancer;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Thyroid cancer, anaplastic;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;not provided
NM_000546.5(TP53):c.742C>T (p.Arg248Trp)	TP53	17	7674221	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Brainstem glioma;Chronic lymphocytic leukemia;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni syndrome 1;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Medulloblastoma;Multiple myeloma;Myelodysplastic syndrome;Neoplasm;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Small cell lung cancer;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Squamous cell carcinoma of the skin;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;not provided
NM_000546.5(TP53):c.743G>A (p.Arg248Gln)	TP53	17	7674220	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Brainstem glioma;Chronic lymphocytic leukemia;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni syndrome 1;Li-Fraumeni syndrome 2;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Medulloblastoma;Multiple myeloma;Myelodysplastic syndrome;Neoplasm;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Sarcoma;Small cell lung cancer;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Squamous cell carcinoma of the skin;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;not provided

NM_005343.3(HRAS):c.34G>A (p.Gly12Ser)	HRAS	11	534289	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Carcinoma, Adenoid Cystic;Colorectal Neoplasms;Costello syndrome;Glioblastoma;Hepatocellular carcinoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Multiple myeloma;Myelodysplastic syndrome;Myopathy, congenital, with excess of muscle spindles;Nasopharyngeal Neoplasms;Neoplasm of breast;Neoplasm of the thyroid gland;Nevus sebaceous;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Papillary renal cell carcinoma, sporadic;Rasopathy;Squamous cell carcinoma of the head and neck;Squamous cell carcinoma of the skin;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;Uterine cervical neoplasms;not provided
NM_001282386.1(IDH1):c.395G>A (p.Arg132His)	IDH1	2	208248388	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of prostate;Astrocytoma;Brainstem glioma;Carcinoma, Adenoid Cystic;Colorectal Neoplasms;Glioblastoma;Glioblastoma multiforme, somatic;Hepatocellular carcinoma;Malignant melanoma of skin;Medulloblastoma;Multiple myeloma;Myelodysplastic syndrome;Neoplasm of brain;Neoplasm of breast;Oligodendroglioma;Transitional cell carcinoma of the bladder
NM_001282387.1(IDH1):c.394C>T (p.Arg132Cys)	IDH1	2	208248389	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of prostate;Astrocytoma;Brainstem glioma;Carcinoma, Adenoid Cystic;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Malignant melanoma of skin;Medulloblastoma;Multiple myeloma;Myelodysplastic syndrome;Neoplasm of brain;Neoplasm of breast;Transitional cell carcinoma of the bladder
NM_000546.5(TP53):c.422G>A (p.Cys141Tyr)	TP53	17	7675190	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of prostate;Colorectal Neoplasms;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Malignant neoplasm of body of uterus;Multiple myeloma;Neoplasm of brain;Neoplasm of breast;Pancreatic adenocarcinoma;Renal cell carcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck
NM_000546.5(TP53):c.535C>T (p.His179Tyr)	TP53	17	7675077	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of stomach;Carcinoma of gallbladder;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Small cell lung cancer;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Squamous cell carcinoma of the skin;Uterine Carcinosarcoma;not provided
NM_005343.3(HRAS):c.436G>A (p.Ala146Thr)	HRAS	11	533467	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of stomach;Colorectal Neoplasms;Costello syndrome;Multiple myeloma;Uterine cervical neoplasms
NM_005343.3(HRAS):c.437C>T (p.Ala146Val)	HRAS	11	533466	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of stomach;Colorectal Neoplasms;Costello syndrome;Multiple myeloma;Uterine cervical neoplasms
NM_000546.5(TP53):c.856G>A (p.Glu286Lys)	TP53	17	7673764	Acute myeloid leukemia;Adenocarcinoma of lung;Adenocarcinoma of stomach;Colorectal Neoplasms;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Malignant melanoma of skin;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Small cell lung cancer;Squamous cell carcinoma of the head and neck;Squamous cell carcinoma of the skin;Transitional cell carcinoma of the bladder;not provided
NM_175629.2(DNMT3A):c.2644C>T (p.Arg882Cys)	DNMT3A	2	25234374	Acute myeloid leukemia;Adenocarcinoma of lung;Myelodysplastic syndrome
NM_175629.2(DNMT3A):c.2645G>A (p.Arg882His)	DNMT3A	2	25234373	Acute myeloid leukemia;Adenocarcinoma of lung;Myelodysplastic syndrome;not provided
NM_002524.4(NRAS):c.35G>A (p.Gly12Asp)	NRAS	1	114716126	Acute myeloid leukemia;Adenocarcinoma of stomach;Colorectal Neoplasms;Epidermal nevus;Juvenile myelomonocytic leukemia;Malignant melanoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Multiple myeloma;Myelodysplastic syndrome;Non-small cell lung cancer;not provided

NM_002524.4(NRAS):c.38G>A (p.Gly13Asp)	NRAS	1	114716123	Acute myeloid leukemia;Adenocarcinoma of stomach;Colorectal Neoplasms;Juvenile myelomonocytic leukemia;Malignant lymphoma, non-Hodgkin;Malignant melanoma;Malignant melanoma of skin;Medulloblastoma;Multiple myeloma;Myelodysplastic syndrome;Noonan syndrome 6;RAS-associated autoimmune leukoproliferative disorder;Transitional cell carcinoma of the bladder;not provided
NM_002524.4(NRAS):c.34G>A (p.Gly12Ser)	NRAS	1	114716127	Acute myeloid leukemia;Adenocarcinoma of stomach;Colorectal Neoplasms;Malignant melanoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Multiple myeloma;Myelodysplastic syndrome;Non-small cell lung cancer;Rasopathy;not provided
NM_001289910.1(IDH2):c.359G>A (p.Arg120Lys)	IDH2	15	90088606	Acute myeloid leukemia;Brainstem glioma;Colorectal Neoplasms;Hepatocellular carcinoma;Myelodysplastic syndrome;Neoplasm of brain
NM_033360.3(KRAS):c.38G>A (p.Gly13Asp)	KRAS	12	25245347	Acute myeloid leukemia;Breast adenocarcinoma;Colorectal Neoplasms;Juvenile myelomonocytic leukemia;Neoplasm of the thyroid gland;Non-small cell lung cancer;Ovarian Neoplasms;RAS-associated autoimmune leukoproliferative disorder
NM_004985.4(KRAS):c.35G>A (p.Gly12Asp)	KRAS	12	25245350	Acute myeloid leukemia;Carcinoma of pancreas;Colorectal Neoplasms;Epidermal nevus;Epidermal nevus syndrome;Juvenile myelomonocytic leukemia;Lung cancer;Neoplasm of ovary;Neoplasm of stomach;Neoplasm of the thyroid gland;Nevus sebaceous;Non-small cell lung cancer;Ovarian Neoplasms;RAS-associated autoimmune leukoproliferative disorder;not provided
NM_156039.3(CSF3R):c.1853C>T (p.Thr618Ile)	CSF3R	1	36467833	Acute myeloid leukemia;Chronic myeloid leukemia;Early T cell progenitor acute lymphoblastic leukemia
NM_002168.3(IDH2):c.419G>A (p.Arg140Gln)	IDH2	15	90088702	Acute myeloid leukemia;Colorectal Neoplasms;D-2-hydroxyglutaric aciduria 2;Multiple myeloma;Myelodysplastic syndrome;Squamous cell carcinoma of the head and neck;not provided
NM_002168.3(IDH2):c.418C>T (p.Arg140Trp)	IDH2	15	90088703	Acute myeloid leukemia;Colorectal Neoplasms;Multiple myeloma;Myelodysplastic syndrome;Squamous cell carcinoma of the head and neck
NM_001145661.1(GATA2):c.1192C>T (p.Arg398Trp)	GATA2	3	128481270	Acute myeloid leukemia;Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency
NM_001145661.1(GATA2):c.1061C>T (p.Thr354Met)	GATA2	3	128481901	Acute myeloid leukemia;Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency;Leukemia, acute myeloid, susceptibility to;Myelodysplastic syndrome
NM_000157.3(GBA):c.27+1G>A	GBA	1	155241085	Acute neuronopathic Gaucher's disease;Gaucher disease;Gaucher's disease, type 1
NM_000157.3(GBA):c.1504C>T (p.Arg502Cys)	GBA	1	155235196	Acute neuronopathic Gaucher's disease;Gaucher disease;Gaucher's disease, type 1;Parkinson disease, late-onset;Subacute neuronopathic Gaucher's disease;not provided
NM_001005741.2(GBA):c.1085C>T (p.Thr362Ile)	GBA	1	155236384	Acute neuronopathic Gaucher's disease;Gaucher's disease, type 1
NM_001005741.2(GBA):c.1090G>A (p.Gly364Arg)	GBA	1	155236379	Acute neuronopathic Gaucher's disease;Gaucher's disease, type 1;not provided
NM_152906.6(TANGO2):c.460G>A (p.Gly154Arg)	TANGO2	22	20061538	Acute rhabdomyolysis;Cardiac arrhythmia;Episodic flaccid weakness;Intellectual functioning disability;Seizures
NM_152906.6(TANGO2):c.605+1G>A	TANGO2	22	20061684	Acute rhabdomyolysis;Cardiac arrhythmia;Episodic flaccid weakness;Intellectual functioning disability;Seizures
NM_014049.4(ACAD9):c.1249C>T (p.Arg417Cys)	ACAD9	3	128906220	Acyl-CoA dehydrogenase family, member 9, deficiency of
NM_014049.4(ACAD9):c.797G>A (p.Arg266Gln)	ACAD9	3	128899450	Acyl-CoA dehydrogenase family, member 9, deficiency of
NR_033426.1(ACAD9):n.887C>T	ACAD9	3	128896491	Acyl-CoA dehydrogenase family, member 9, deficiency of
NM_014049.4(ACAD9):c.1594C>T (p.Arg532Trp)	ACAD9	3	128910051	Acyl-CoA dehydrogenase family, member 9, deficiency of;not provided
NM_020754.3(ARHGAP31):c.2047C>T (p.Gln683Ter)	ARHGAP31	3	119413976	Adams-Oliver syndrome 1
NM_017617.4(NOTCH1):c.5965G>A (p.Asp1989Asn)	NOTCH1	9	136499229	Adams-Oliver syndrome 1;Adams-Oliver syndrome 5
NM_020812.3(DOCK6):c.3154G>A (p.Glu1052Lys)	DOCK6	19	11222821	Adams-Oliver syndrome 2
NM_001278689.1(EOGT):c.1130G>A (p.Arg377Gln)	EOGT	3	68987467	Adams-Oliver syndrome 4
NM_017617.4(NOTCH1):c.4487G>A (p.Cys1496Tyr)	NOTCH1	9	136505409	Adams-Oliver syndrome 5

NM_017617.4(NOTCH1):c.1343G>A (p.Arg448Gln)	NOTCH1	9	136517850	Adams-Oliver syndrome 5	
NM_019074.3(DLL4):c.556C>T (p.Arg186Cys)	DLL4	15	40931664	Adams-Oliver syndrome;Adams-Oliver syndrome 6	
NM_019074.3(DLL4):c.1169G>A (p.Cys390Tyr)	DLL4	15	40935046	Adams-Oliver syndrome;Adams-Oliver syndrome 6	
NM_019074.3(DLL4):c.1660C>T (p.Gln554Ter)	DLL4	15	40936647	Adams-Oliver syndrome;Adams-Oliver syndrome 6	
NM_019074.3(DLL4):c.1672C>T (p.Arg558Ter)	DLL4	15	40936659	Adams-Oliver syndrome;Adams-Oliver syndrome 6	
NM_000033.3(ABCD1):c.1817C>T (p.Ser606Leu)	ABCD1	X	153743023	Addison's disease;Adrenoleukodystrophy	
NM_000485.2(APRT):c.294G>A (p.Trp98Ter)	APRT	16	88810450	Adenine phosphoribosyltransferase deficiency	
NM_006218.3(PIK3CA):c.3139C>T (p.His1047Tyr)	PIK3CA	3	179234296	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Adrenocortical Carcinoma;Brainstem glioma;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Malignant tumor of floor of mouth;Medulloblastoma;Megalencephaly cutis marmorata telangiectatica congenita;Neoplasm of brain;Neoplasm of breast;Non-small cell lung cancer;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Renal cell carcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;Uterine cervical neoplasms	
NM_000546.5(TP53):c.638G>A (p.Arg213Gln)	TP53	17	7674893	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Adrenocortical Carcinoma;Carcinoma, Adenoid Cystic;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni syndrome 1;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Nasopharyngeal Neoplasms;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Renal cell carcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Squamous cell carcinoma of the skin;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;not provided;not specified	
NM_006218.3(PIK3CA):c.1633G>A (p.Glu545Lys)	PIK3CA	3	179218303	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Brainstem glioma;Breast adenocarcinoma;Carcinoma of colon;Carcinoma of gallbladder;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Keratosis, seborrheic;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Megalencephaly cutis marmorata telangiectatica congenita;Nasopharyngeal Neoplasms;Neoplasm of brain;Neoplasm of breast;Neoplasm of stomach;Non-small cell lung cancer;Oesophageal carcinoma;Ovarian Neoplasms;Ovarian Serous Cystadenocarcinoma;Ovarian epithelial cancer;Pancreatic adenocarcinoma;Papillary renal cell carcinoma, sporadic;Renal cell carcinoma;Sarcoma;Small cell lung cancer;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;Uterine cervical neoplasms	
NM_000546.5(TP53):c.734G>A (p.Gly245Asp)	TP53	17	7674229	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Brainstem glioma;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni syndrome 1;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma	

NM_005163.2(AKT1):c.49G>A (p.Glu17Lys)	AKT1	14	104780214	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Breast adenocarcinoma;Carcinoma of colon;Colorectal Neoplasms;Endometrial Endometrioid Adenocarcinoma, Variant with Squamous Differentiation;Hepatocellular carcinoma;Malignant melanoma of skin;Meningeal Neoplasms;Neoplasm of breast;Neoplasm of ovary;Neoplasm of the thyroid gland;Non-small cell lung cancer;Osteosarcoma;Prostatic Neoplasms;Proteus syndrome;Small cell lung cancer;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine cervical neoplasms
NM_006218.3(PIK3CA):c.1624G>A (p.Glu542Lys)	PIK3CA	3	179218294	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Colorectal Neoplasms;Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi;Glioblastoma;Hepatocellular carcinoma;Malignant neoplasm of body of uterus;Neoplasm of brain;Neoplasm of breast;Neoplasm of ovary;Non-small cell lung cancer;Oesophageal carcinoma;Ovarian Neoplasms;Papillary renal cell carcinoma, sporadic;Small cell lung cancer;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine cervical neoplasms;not provided
NM_001904.3(CTNNB1):c.98C>T (p.Ser33Phe)	CTNNB1	3	41224610	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Colorectal Neoplasms;Hepatocellular carcinoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Medulloblastoma;Oesophageal carcinoma;Pancreatic adenocarcinoma;Pilomatrixoma;Transitional cell carcinoma of the bladder
NM_005359.5(SMAD4):c.1157G>A (p.Gly386Asp)	SMAD4	18	51067036	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Colorectal Neoplasms;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;Oesophageal carcinoma;Pancreatic adenocarcinoma
NM_001904.3(CTNNB1):c.110C>T (p.Ser37Phe)	CTNNB1	3	41224622	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Hepatocellular carcinoma;Malignant melanoma;Malignant neoplasm of body of uterus;Medulloblastoma;Oesophageal carcinoma;Ovarian Neoplasms;Pilomatrixoma;Transitional cell carcinoma of the bladder;Uterine cervical neoplasms
NM_001904.3(CTNNB1):c.134C>T (p.Ser45Phe)	CTNNB1	3	41224646	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adrenocortical Carcinoma;Colorectal Neoplasms;Disease;Hepatocellular carcinoma;Malignant melanoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Transitional cell carcinoma of the bladder
NM_001904.3(CTNNB1):c.122C>T (p.Thr41Ile)	CTNNB1	3	41224634	Adenocarcinoma of lung;Adenocarcinoma of prostate;Adrenocortical Carcinoma;Colorectal Neoplasms;Hepatocellular carcinoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Pancreatic adenocarcinoma;Pilomatrixoma
NM_004333.4(BRAF):c.1406G>A (p.Gly469Glu)	BRAF	7	140781602	Adenocarcinoma of lung;Adenocarcinoma of prostate;Cardio-facio-cutaneous syndrome;Cardiofaciocutaneous syndrome 1;Colorectal Neoplasms;Malignant melanoma;Malignant melanoma of skin;Multiple myeloma;Rasopathy;Squamous cell carcinoma of lung;Squamous cell carcinoma of the skin;Transitional cell carcinoma of the bladder;not provided
NM_000516.5(GNAS):c.601C>T (p.Arg201Cys)	GNAS	20	58909365	Adenocarcinoma of lung;Adenocarcinoma of stomach;Adrenocortical Carcinoma;Colorectal Neoplasms;Cushing's syndrome;Hepatocellular carcinoma;Malignant melanoma of skin;McCune-Albright syndrome;Neoplasm;Neoplasm of breast;Pancreatic adenocarcinoma;Sex cord-stromal tumor;Somatotroph adenoma;Squamous cell carcinoma of the head and neck;Uterine cervical neoplasms
NM_000516.5(GNAS):c.602G>A (p.Arg201His)	GNAS	20	58909366	Adenocarcinoma of lung;Adenocarcinoma of stomach;Adrenocortical Carcinoma;Colorectal Neoplasms;Cushing's syndrome;Hepatocellular carcinoma;Malignant melanoma of skin;McCune-Albright syndrome;Neoplasm;Neoplasm of breast;Pancreatic adenocarcinoma;Sex cord-stromal tumor;Somatotroph adenoma;Squamous cell carcinoma of the head and neck;Uterine cervical neoplasms

NM_001904.3(CTNNB1):c.101G>A (p.Gly34Glu)	CTNNB1	3	41224613	Adenocarcinoma of lung;Adenocarcinoma of stomach;Adrenocortical Carcinoma;Hepatocellular carcinoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Medulloblastoma;Ovarian Neoplasms;Pilomatrixoma;Squamous cell carcinoma of the head and neck
NM_000546.5(TP53):c.451C>T (p.Pro151Ser)	TP53	17	7675161	Adenocarcinoma of lung;Adenocarcinoma of stomach;Breast adenocarcinoma;Carcinoma, Adenoid Cystic;Colorectal Neoplasms;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Multiple myeloma;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;not provided
NM_000546.5(TP53):c.725G>A (p.Cys242Tyr)	TP53	17	7674238	Adenocarcinoma of lung;Adenocarcinoma of stomach;Chronic lymphocytic leukemia;Glioblastoma;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni-like syndrome;Neoplasm of breast;Oesophageal carcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma
NM_005359.5(SMAD4):c.1081C>T (p.Arg361Cys)	SMAD4	18	51065548	Adenocarcinoma of lung;Adenocarcinoma of stomach;Colorectal Neoplasms;Juvenile polyposis syndrome;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;Neoplasm of breast;Oesophageal carcinoma;Pancreatic adenocarcinoma;Squamous cell carcinoma of the head and neck;Uterine cervical neoplasms;not provided
NM_002880.3(RAF1):c.770C>T (p.Ser257Leu)	RAF1	3	12604200	Adenocarcinoma of lung;Adenocarcinoma of stomach;Colorectal Neoplasms;LEOPARD syndrome 2;Malignant melanoma of skin;Noonan syndrome;Noonan syndrome 5;Noonan syndrome with multiple lentigines;Rasopathy;not provided
NM_005359.5(SMAD4):c.1082G>A (p.Arg361His)	SMAD4	18	51065549	Adenocarcinoma of lung;Adenocarcinoma of stomach;Colorectal Neoplasms;Neoplasm of breast;Oesophageal carcinoma;Pancreatic adenocarcinoma;Squamous cell carcinoma of the head and neck;Uterine cervical neoplasms
NM_000546.5(TP53):c.824G>A (p.Cys275Tyr)	TP53	17	7673796	Adenocarcinoma of lung;Adrenocortical Carcinoma;Chronic lymphocytic leukemia;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Li-Fraumeni syndrome;Malignant melanoma of skin;Multiple myeloma;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Renal cell carcinoma;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;not provided
NM_004333.4(BRAF):c.1780G>A (p.Asp594Asn)	BRAF	7	140753355	Adenocarcinoma of lung;Adrenocortical Carcinoma;Chronic lymphocytic leukemia;Colorectal Neoplasms;Malignant melanoma;Malignant melanoma of skin;Multiple myeloma;Neoplasm of brain;Non-small cell lung cancer;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder
NM_000142.4(FGFR3):c.742C>T (p.Arg248Cys)	FGFR3	4	1801837	Adenocarcinoma of lung;Bell-shaped thorax;Bowed humerus;Carcinoma;Disproportionate short-limb short stature;Epidermal nevus;Femoral bowing;Growth delay;Keratosis, seborrheic;Lethal short-limbed short stature;Lower limb undergrowth;Multiple myeloma;Narrow chest;Short ribs;Short stature;Skeletal dysplasia;Skeletal dysplasia with acanthosis nigricans;Small for gestational age;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Thanatophoric dysplasia type 1;Transitional cell carcinoma of the bladder;Upper limb undergrowth;not provided

NM_000546.5(TP53):c.722C>T (p.Ser241Phe)	TP53	17	7674241	Adenocarcinoma of lung;Brainstem glioma;Carcinoma of gallbladder;Colorectal Neoplasms;Glioblastoma;Hepatoblastoma;Hereditary cancer-predisposing syndrome;Malignant lymphoma, non-Hodgkin;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Osteosarcoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Papillary renal cell carcinoma, sporadic;Renal cell carcinoma;Squamous cell carcinoma of the head and neck;Squamous cell carcinoma of the skin;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma
NM_006218.3(PIK3CA):c.2176G>A (p.Glu726Lys)	PIK3CA	3	179221146	Adenocarcinoma of lung;Brainstem glioma;Colorectal Neoplasms;Neoplasm of breast;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine cervical neoplasms;not provided
NM_000038.5(APC):c.3340C>T (p.Arg1114Ter)	APC	5	112838934	Adenocarcinoma of lung;Colorectal Neoplasms;Familial adenomatous polyposis 1
NM_000546.5(TP53):c.580C>T (p.Leu194Phe)	TP53	17	7674951	Adenocarcinoma of lung;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Neoplasm of brain;Neoplasm of breast;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine cervical neoplasms;not provided
NM_000546.5(TP53):c.833C>T (p.Pro278Leu)	TP53	17	7673787	Adenocarcinoma of lung;Colorectal Neoplasms;Hereditary cancer-predisposing syndrome;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Multiple myeloma;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Squamous cell carcinoma of the skin
NM_000075.3(CDK4):c.70C>T (p.Arg24Cys)	CDK4	12	57751648	Adenocarcinoma of lung;Cutaneous malignant melanoma 3;Hereditary cutaneous melanoma;Malignant melanoma of skin;Multiple myeloma;not provided
NM_000455.4(STK11):c.580G>A (p.Asp194Asn)	STK11	19	1220488	Adenocarcinoma of lung;Neoplasm;Peutz-Jeghers syndrome
NM_000314.6(PTEN):c.389G>A (p.Arg130Gln)	PTEN	10	87933148	Adenocarcinoma of prostate;Adenocarcinoma of stomach;Colorectal Neoplasms;Cowden syndrome 1;Glioblastoma;Hereditary cancer-predisposing syndrome;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Neoplasm;Neoplasm of breast;Ovarian Neoplasms;PTEN hamartoma tumor syndrome;Renal cell carcinoma;Small cell lung cancer;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Uterine Carcinosarcoma;Uterine cervical neoplasms;not provided
NM_006218.3(PIK3CA):c.353G>A (p.Gly118Asp)	PIK3CA	3	179199690	Adenocarcinoma of prostate;Adenocarcinoma of stomach;Cowden syndrome 5;Glioblastoma;Malignant neoplasm of body of uterus;Neoplasm of brain;Neoplasm of breast;Neoplasm of the thyroid gland;Pancreatic adenocarcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Uterine cervical neoplasms
NM_004448.3(ERBB2):c.2524G>A (p.Val842Ile)	ERBB2	17	39725079	Adenocarcinoma of stomach;Carcinoma of gallbladder;Colorectal Neoplasms;Malignant neoplasm of body of uterus;Neoplasm of breast;Pancreatic adenocarcinoma;Uterine Carcinosarcoma
NM_006218.3(PIK3CA):c.3129G>A (p.Met1043Ile)	PIK3CA	3	179234286	Adenocarcinoma of stomach;Carcinoma, Adenoid Cystic;Colorectal Neoplasms;Glioblastoma;Malignant neoplasm of body of uterus;Neoplasm of brain;Neoplasm of breast;Neoplasm of the thyroid gland;Non-small cell lung cancer;PIK3CA related overgrowth spectrum;Pancreatic adenocarcinoma;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder
NM_001005862.2(ERBB2):c.2236G>A (p.Gly746Ser)	ERBB2	17	39724744	Adenocarcinoma of stomach;Neoplasm;Neoplasm of stomach

NM_000546.5(TP53):c.733G>A (p.Gly245Ser)	TP53	17	7674230	Adenocarcinoma;Adenocarcinoma of lung;Adenocarcinoma of prostate;Adenocarcinoma of stomach;Brainstem glioma;Colorectal Neoplasms;Glioblastoma;Hepatocellular carcinoma;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni syndrome 1;Li-Fraumeni-like syndrome;Neoplasm;Neoplasm of brain;Neoplasm of breast;Oesophageal carcinoma;Ovarian Serous Cystadenocarcinoma;Pancreatic adenocarcinoma;Squamous cell carcinoma of lung;Squamous cell carcinoma of the head and neck;Transitional cell carcinoma of the bladder;Uterine Carcinosarcoma;not provided
NM_000298.5(PKLR):c.110G>A (p.Gly37Glu)	PKLR	1	155300271	Adenosine triphosphate, elevated, of erythrocytes
NM_000476.2(AK1):c.382C>T (p.Arg128Trp)	AK1	9	127868455	Adenylate kinase deficiency, hemolytic anemia due to
NM_000476.2(AK1):c.319C>T (p.Arg107Ter)	AK1	9	127871828	Adenylate kinase deficiency, hemolytic anemia due to
NM_000476.2(AK1):c.118G>A (p.Gly40Arg)	AK1	9	127872779	Adenylate kinase deficiency, hemolytic anemia due to
NM_000476.2(AK1):c.190G>A (p.Gly64Arg)	AK1	9	127872707	Adenylate kinase deficiency, hemolytic anemia due to
NM_000026.3(ADSL):c.907C>T (p.Arg303Cys)	ADSL	22	40361532	Adenylosuccinate lyase deficiency
NM_000026.3(ADSL):c.1277G>A (p.Arg426His)	ADSL	22	40364965	Adenylosuccinate lyase deficiency;not provided
NM_000026.3(ADSL):c.569G>A (p.Arg190Gln)	ADSL	22	40358950	Adenylosuccinate lyase deficiency;not provided
NM_000888.4(ITGB6):c.898G>A (p.Glu300Lys)	ITGB6	2	160172592	Adolescent alopeciam dentogingival abnormalitites and intellectual disability
NM_153240.4(NPHP3):c.2994G>A (p.Trp998Ter)	NPHP3	3	132688781	Adolescent nephronophthisis
NM_153240.4(NPHP3):c.1729C>T (p.Arg577Ter)	NPHP3	3	132700348	Adolescent nephronophthisis;Meckel syndrome type 7;Renal-hepatic-pancreatic dysplasia;not provided
NM_153240.4(NPHP3):c.2104C>T (p.Arg702Ter)	NPHP3	3	132696798	Adolescent nephronophthisis;Nephronophthisis
NM_153240.4(NPHP3):c.3373C>T (p.Arg1125Ter)	NPHP3	3	132684751	Adolescent nephronophthisis;Renal-hepatic-pancreatic dysplasia;not provided
NM_000781.2(CYP11A1):c.1057C>T (p.Arg353Trp)	CYP11A1	15	74339687	Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete
NM_000781.2(CYP11A1):c.566C>T (p.Ala189Val)	CYP11A1	15	74345103	Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete
NM_000781.2(CYP11A1):c.1076C>T (p.Ala359Val)	CYP11A1	15	74339668	Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete
NM_000546.5(TP53):c.1010G>A (p.Arg337His)	TP53	17	7670699	Adrenocortical carcinoma, pediatric;Breast cancer;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;not provided
NM_002734.4(PRKAR1A):c.892-1G>A	PRKAR1A	17	68529919	Adrenocortical tumor, somatic
NM_002070.3(GNAI2):c.535C>T (p.Arg179Cys)	GNAI2	3	50256262	Adrenocortical tumor, somatic;Granulosa cell tumor of the ovary;Thecoma, somatic
NM_002070.3(GNAI2):c.536G>A (p.Arg179His)	GNAI2	3	50256263	Adrenocortical tumor, somatic;Granulosa cell tumor of the ovary;Thecoma, somatic
NM_000033.3(ABCD1):c.871G>A (p.Glu291Lys)	ABCD1	X	153726137	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.796G>A (p.Gly266Arg)	ABCD1	X	153726062	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1202G>A (p.Arg401Gln)	ABCD1	X	153736232	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1252C>T (p.Arg418Trp)	ABCD1	X	153736372	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1390C>T (p.Arg464Ter)	ABCD1	X	153736510	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1544C>T (p.Ser515Phe)	ABCD1	X	153740147	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1552C>T (p.Arg518Trp)	ABCD1	X	153740155	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1850G>A (p.Arg617His)	ABCD1	X	153743056	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1849C>T (p.Arg617Cys)	ABCD1	X	153743055	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.421G>A (p.Ala141Thr)	ABCD1	X	153725687	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1396C>T (p.Gln466Ter)	ABCD1	X	153737159	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1553G>A (p.Arg518Gln)	ABCD1	X	153740156	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1679C>T (p.Pro560Leu)	ABCD1	X	153740618	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1771C>T (p.Arg591Trp)	ABCD1	X	153740710	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1802G>A (p.Trp601Ter)	ABCD1	X	153743008	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.346G>A (p.Gly116Arg)	ABCD1	X	153725612	Adrenoleukodystrophy

NM_000033.3(ABCD1):c.406C>T (p.Gln136Ter)	ABCD1	X	153725672	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1661G>A (p.Arg554His)	ABCD1	X	153740600	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1825G>A (p.Glu609Lys)	ABCD1	X	153743031	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1288C>T (p.Gln430Ter)	ABCD1	X	153736408	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1781-1G>A	ABCD1	X	153742986	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.529C>T (p.Gln177Ter)	ABCD1	X	153725795	Adrenoleukodystrophy
NM_000033.3(ABCD1):c.1866-10G>A	ABCD1	X	153743211	Adrenoleukodystrophy;not provided
NM_000478.5(ALPL):c.571G>A (p.Glu191Lys)	ALPL	1	21564139	Adult hypophosphatasia;Childhood hypophosphatasia;Hypophosphatasia;Infantile hypophosphatasia;not provided
NM_000478.5(ALPL):c.346G>A (p.Ala116Thr)	ALPL	1	21563158	Adult hypophosphatasia;Childhood hypophosphatasia;Infantile hypophosphatasia;Odontohypophosphatasia;not provided
NM_198129.2(LAMA3):c.8962C>T (p.Gln2988Ter)	LAMA3	18	23939322	Adult junctional epidermolysis bullosa
NM_000228.2(LAMB3):c.628G>A (p.Glu210Lys)	LAMB3	1	209633070	Adult junctional epidermolysis bullosa
NM_000228.2(LAMB3):c.628+42G>A	LAMB3	1	209633028	Adult junctional epidermolysis bullosa
NM_000228.2(LAMB3):c.629-1G>A	LAMB3	1	209632777	Adult junctional epidermolysis bullosa
NM_005562.2(LAMC2):c.733C>T (p.Arg245Ter)	LAMC2	1	183222181	Adult junctional epidermolysis bullosa
NM_000213.4(ITGB4):c.2792G>A (p.Gly931Asp)	ITGB4	17	75742591	Adult junctional epidermolysis bullosa
NM_000494.3(COL17A1):c.3067C>T (p.Gln1023Ter)	COL17A1	10	104038409	Adult junctional epidermolysis bullosa
NM_000494.3(COL17A1):c.1898G>A (p.Gly633Asp)	COL17A1	10	104053072	Adult junctional epidermolysis bullosa
NM_000494.3(COL17A1):c.433C>T (p.Arg145Ter)	COL17A1	10	104072062	Adult junctional epidermolysis bullosa
NM_000494.3(COL17A1):c.3676C>T (p.Arg1226Ter)	COL17A1	10	104034711	Adult junctional epidermolysis bullosa;Epidermolysis bullosa, junctional, localisata variant
NM_000228.2(LAMB3):c.1903C>T (p.Arg635Ter)	LAMB3	1	209625721	Adult junctional epidermolysis bullosa;Junctional epidermolysis bullosa gravis of Herlitz;not provided
NM_017882.2(CLN6):c.308G>A (p.Arg103Gln)	CLN6	15	68211853	Adult neuronal ceroid lipofuscinosis;not provided
NM_017882.2(CLN6):c.139C>T (p.Leu47Phe)	CLN6	15	68218595	Adult neuronal ceroid lipofuscinosis;not provided
NM_175073.2(APTX):c.617C>T (p.Pro206Leu)	APTX	9	32984784	Adult onset ataxia with oculomotor apraxia
NM_175073.2(APTX):c.837G>A (p.Trp279Ter)	APTX	9	32974495	Adult onset ataxia with oculomotor apraxia;not provided
NM_003722.4(TP63):c.1010G>A (p.Arg337Gln)	TP63	3	189868597	ADULT syndrome
NM_003722.4(TP63):c.518G>A (p.Gly173Asp)	TP63	3	189808465	ADULT syndrome
NM_003722.4(TP63):c.797G>A (p.Arg266Gln)	TP63	3	189866712	ADULT syndrome;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3;not provided
NM_001031726.3(C19orf12):c.205G>A (p.Gly69Arg)	C19orf12	19	29702966	Adult-onset night blindness;Dystonia;Mental deterioration;Neurodegeneration with brain iron accumulation 4;Peripheral visual field loss;Tremor
NM_000204.4(CFI):c.728G>A (p.Gly243Asp)	CFI	4	109760567	Afibrinogenemia
NM_021870.2(FGG):c.78+5G>A	FGG	4	154612527	Afibrinogenemia, congenital
NM_021870.2(FGG):c.307+5G>A	FGG	4	154612013	Afibrinogenemia, congenital
NM_005141.4(FGB):c.1289G>A (p.Gly430Asp)	FGB	4	154570463	Afibrinogenemia, congenital
NM_005141.4(FGB):c.958+13C>T	FGB	4	154569320	Afibrinogenemia, congenital
NM_005141.4(FGB):c.139C>T (p.Arg47Ter)	FGB	4	154565832	Afibrinogenemia, congenital
NM_000508.4(FGA):c.502C>T (p.Arg168Ter)	FGA	4	154587520	Afibrinogenemia, congenital
NM_020070.3(IGLL1):c.64C>T (p.Gln22Ter)	IGLL1	22	23580127	Agammaglobulinemia 2, autosomal recessive
NM_020070.3(IGLL1):c.425C>T (p.Pro142Leu)	IGLL1	22	23573483	Agammaglobulinemia 2, autosomal recessive
NM_001039933.2(CD79B):c.412G>A (p.Gly138Ser)	CD79B	17	63930095	Agammaglobulinemia 6, autosomal recessive
NM_001039933.2(CD79B):c.241C>T (p.Gln81Ter)	CD79B	17	63930266	Agammaglobulinemia 6, autosomal recessive
NM_181523.2(PIK3R1):c.893G>A (p.Trp298Ter)	PIK3R1	5	68280983	Agammaglobulinemia 7, autosomal recessive
NM_001136139.2(TCF3):c.1663G>A (p.Glu555Lys)	TCF3	19	1612357	Agammaglobulinemia 8, autosomal dominant
NC_000014.9:g.105855107C>T	IGHM	14	105855107	Agammaglobulinemia, non-Bruton type

NM_153704.5(TMEM67):c.1321C>T (p.Arg441Cys)	TMEM67	8	93786255	Agenesis of cerebellar vermis;Joubert syndrome 6;Oligohydramnios;Renal cyst
NM_020732.3(ARID1B):c.1612C>T (p.Gln538Ter)	ARID1B	6	156829296	Agenesis of corpus callosum;Global developmental delay;Hypertrichosis;Nail dysplasia;Neonatal hypotonia
NM_000099.3(CST3):c.73G>A (p.Ala25Thr)	CST3	20	23637790	Age-related macular degeneration 11
NM_000350.2(ABCA4):c.1222C>T (p.Arg408Ter)	ABCA4	1	94079339	Age-related macular degeneration 2;Cone-rod dystrophy 3;Retinitis pigmentosa 19;Stargardt disease 1;not provided
NM_006329.3(FBLN5):c.212G>A (p.Arg71Gln)	FBLN5	14	91937114	Age-related macular degeneration 3
NM_006329.3(FBLN5):c.259C>T (p.Pro87Ser)	FBLN5	14	91937067	Age-related macular degeneration 3
NM_006329.3(FBLN5):c.1051C>T (p.Arg351Trp)	FBLN5	14	91877621	Age-related macular degeneration 3
NM_006329.3(FBLN5):c.1087G>A (p.Ala363Thr)	FBLN5	14	91877585	Age-related macular degeneration 3
NM_006329.3(FBLN5):c.1235G>A (p.Gly412Glu)	FBLN5	14	91870336	Age-related macular degeneration 3
NM_006329.3(FBLN5):c.1117C>T (p.Arg373Cys)	FBLN5	14	91877555	Age-related macular degeneration 3
NM_000186.3(CFH):c.3628C>T (p.Arg1210Cys)	CFH	1	196747245	Age-related macular degeneration 4;Atypical hemolytic-uremic syndrome 1;Factor H deficiency
NM_032753.3(RAX2):c.260G>A (p.Arg87Gln)	RAX2	19	3770916	Age-related macular degeneration 6
NM_002775.4(HTRA1):c.-625G>A	HTRA1	10	122461028	Age-related macular degeneration 7;Susceptibility to neovascular type of age-related macular degeneration
NM_016381.5(TREX1):c.794G>A (p.Trp265Ter)	TREX1	3	48467284	Aicardi Goutieres syndrome 1
NM_016381.5(TREX1):c.763G>A (p.Asp255Asn)	TREX1	3	48467253	Aicardi Goutieres syndrome 1;Aicardi Goutieres syndrome 1, autosomal dominant
NM_033629.4(TREX1):c.52G>A (p.Asp18Asn)	TREX1	3	48466707	Aicardi Goutieres syndrome 1;Aicardi Goutieres syndrome 1, autosomal dominant;Chilblain lupus 1;not provided
NM_033629.4(TREX1):c.490C>T (p.Arg164Ter)	TREX1	3	48467145	Aicardi Goutieres syndrome 1;not provided
NM_032193.3(RNASEH2C):c.205C>T (p.Arg69Trp)	RNASEH2C	11	65720385	Aicardi Goutieres syndrome 3;not provided
NM_006397.2(RNASEH2A):c.109G>A (p.Gly37Ser)	RNASEH2A	19	12806782	Aicardi Goutieres syndrome 4
NM_006397.2(RNASEH2A):c.75C>T (p.Arg25=)	RNASEH2A	19	12806748	Aicardi Goutieres syndrome 4
NM_006397.2(RNASEH2A):c.704G>A (p.Arg235Gln)	RNASEH2A	19	12813149	Aicardi Goutieres syndrome 4
NM_006397.2(RNASEH2A):c.69G>A (p.Val23=)	RNASEH2A	19	12806742	Aicardi Goutieres syndrome 4
NM_006397.2(RNASEH2A):c.556C>T (p.Arg186Trp)	RNASEH2A	19	12810323	Aicardi Goutieres syndrome 4
NM_006397.2(RNASEH2A):c.322C>T (p.Arg108Trp)	RNASEH2A	19	12807328	Aicardi Goutieres syndrome 4
NM_006397.2(RNASEH2A):c.719C>T (p.Thr240Met)	RNASEH2A	19	12813164	Aicardi Goutieres syndrome 4
NM_006397.2(RNASEH2A):c.872G>A (p.Arg291His)	RNASEH2A	19	12813438	Aicardi Goutieres syndrome 4
NM_015474.3(SAMHD1):c.625G>A (p.Gly209Ser)	SAMHD1	20	36930760	Aicardi Goutieres syndrome 5
NM_015474.3(SAMHD1):c.433C>T (p.Arg145Ter)	SAMHD1	20	36935105	Aicardi Goutieres syndrome 5
NM_015474.3(SAMHD1):c.490C>T (p.Arg164Ter)	SAMHD1	20	36935048	Aicardi Goutieres syndrome 5
NM_015474.3(SAMHD1):c.445C>T (p.Gln149Ter)	SAMHD1	20	36935093	Aicardi Goutieres syndrome 5
NM_015474.3(SAMHD1):c.1642C>T (p.Gln548Ter)	SAMHD1	20	36897926	Aicardi Goutieres syndrome 5
NM_015474.3(SAMHD1):c.427C>T (p.Arg143Cys)	SAMHD1	20	36935111	Aicardi Goutieres syndrome 5
NM_015474.3(SAMHD1):c.1324C>T (p.Arg442Ter)	SAMHD1	20	36905450	Aicardi Goutieres syndrome 5
NM_015474.3(SAMHD1):c.428G>A (p.Arg143His)	SAMHD1	20	36935110	Aicardi Goutieres syndrome 5
NM_015474.3(SAMHD1):c.434G>A (p.Arg145Gln)	SAMHD1	20	36935104	Aicardi Goutieres syndrome 5
NM_001111.4(ADAR):c.2675G>A (p.Arg892His)	ADAR	1	154589456	Aicardi-goutieres syndrome 6
NM_001111.4(ADAR):c.2608G>A (p.Ala870Thr)	ADAR	1	154589817	Aicardi-goutieres syndrome 6
NM_001111.4(ADAR):c.1630C>T (p.Arg544Ter)	ADAR	1	154598557	Aicardi-goutieres syndrome 6
NM_001111.4(ADAR):c.3019G>A (p.Gly1007Arg)	ADAR	1	154588125	Aicardi-goutieres syndrome 6;Symmetrical dyschromatosis of extremities
NM_022168.3(IFIH1):c.2159G>A (p.Arg720Gln)	IFIH1	2	162276832	Aicardi-goutieres syndrome 7
NM_022168.3(IFIH1):c.1483G>A (p.Gly495Arg)	IFIH1	2	162281369	Aicardi-goutieres syndrome 7

NM_022168.3(IFIH1):c.1354G>A (p.Ala452Thr)	IFIH1	2	162281498	Aicardi-goutieres syndrome 7	
NM_022168.3(IFIH1):c.1114C>T (p.Leu372Phe)	IFIH1	2	162282558	Aicardi-goutieres syndrome 7	
NM_022168.3(IFIH1):c.2336G>A (p.Arg779His)	IFIH1	2	162273913	Aicardi-goutieres syndrome 7;not provided	
NM_022168.3(IFIH1):c.2335C>T (p.Arg779Cys)	IFIH1	2	162273914	Aicardi-goutieres syndrome 7;not provided	
NM_014336.4(AIPL1):c.834G>A (p.Trp278Ter)	AIPL1	17	6425781	AIPL1-Related Disorders;Leber congenital amaurosis 4;not provided	
NM_013335.3(GMPPA):c.295C>T (p.Arg99Ter)	GMPPA	2	219501903	Alacrima, achalasia, and mental retardation syndrome	
NM_013335.3(GMPPA):c.545G>A (p.Gly182Asp)	GMPPA	2	219504138	Alacrima, achalasia, and mental retardation syndrome	
NM_000214.2(JAG1):c.550C>T (p.Arg184Cys)	JAG1	20	10658612	Alagille syndrome 1	
NM_000214.2(JAG1):c.551G>A (p.Arg184His)	JAG1	20	10658611	Alagille syndrome 1	
NM_000214.2(JAG1):c.1326G>A (p.Trp442Ter)	JAG1	20	10649544	Alagille syndrome 1	
NM_000214.2(JAG1):c.1207C>T (p.Gln403Ter)	JAG1	20	10650274	Alagille syndrome 1	
NM_000214.2(JAG1):c.841C>T (p.Gln281Ter)	JAG1	20	10652513	Alagille syndrome 1	
NM_000214.2(JAG1):c.439C>T (p.Gln147Ter)	JAG1	20	10663963	Alagille syndrome 1	
NM_000214.2(JAG1):c.2122C>T (p.Gln708Ter)	JAG1	20	10645248	Alagille syndrome 1	
NM_000214.2(JAG1):c.2688G>A (p.Trp896Ter)	JAG1	20	10641688	Alagille syndrome 1	
NM_000214.2(JAG1):c.703C>T (p.Arg235Ter)	JAG1	20	10656450	Alagille syndrome 1;not provided	
NM_024408.3(NOTCH2):c.5930-1G>A	NOTCH2	1	119917763	Alagille syndrome 2	
NM_024408.3(NOTCH2):c.1331G>A (p.Cys444Tyr)	NOTCH2	1	119967555	Alagille syndrome 2	
NM_024408.3(NOTCH2):c.1147C>T (p.Pro383Ser)	NOTCH2	1	119968194	Alagille syndrome 2	
NM_024408.3(NOTCH2):c.1180C>T (p.Pro394Ser)	NOTCH2	1	119968161	Alagille syndrome 2	
NM_024408.3(NOTCH2):c.5857C>T (p.Arg1953Cys)	NOTCH2	1	119918478	Alagille syndrome 2	
NM_024408.3(NOTCH2):c.5858G>A (p.Arg1953His)	NOTCH2	1	119918477	Alagille syndrome 2	
NM_024408.3(NOTCH2):c.6007C>T (p.Arg2003Ter)	NOTCH2	1	119917685	Alagille syndrome 2;Hajdu-Cheney syndrome	
NM_005641.3(TAF6):c.136C>T (p.Arg46Cys)	TAF6	7	100114074	ALAZAMI-YUAN SYNDROME;Cornelia de Lange syndrome 1	
NM_205850.2(SLC24A5):c.991G>A (p.Trp197Ter)	SLC24A5	15	48136683	Albinism, oculocutaneous, type VI	
NM_001305581.1(LRMDA):c.664C>T (p.Arg222Ter)	LRMDA	10	76557271	Albinism, oculocutaneous, type VII	
NM_001305581.1(LRMDA):c.256C>T (p.Arg86Ter)	LRMDA	10	76036132	Albinism, oculocutaneous, type VII	
NM_000669.4(ADH1C):c.815G>A (p.Arg272Gln)	ADH1C	4	99342808	Alcohol dependence	
NM_000668.5(ADH1B):c.1108C>T (p.Arg370Cys)	ADH1B	4	99307860	Alcohol dependence	
NM_002055.4(GFAP):c.1193C>T (p.Ser398Phe)	GFAP	17	44908128	Alexander's disease	
NM_002055.4(GFAP):c.1154C>T (p.Ser385Phe)	GFAP	17	44910632	Alexander's disease	
NM_002055.4(GFAP):c.1090G>A (p.Ala364Thr)	GFAP	17	44911273	Alexander's disease	
NM_002055.4(GFAP):c.1073C>T (p.Ala358Val)	GFAP	17	44911290	Alexander's disease	
NM_002055.4(GFAP):c.772C>T (p.Arg258Cys)	GFAP	17	44913277	Alexander's disease	
NM_002055.4(GFAP):c.259G>A (p.Val87Ile)	GFAP	17	44915228	Alexander's disease	
NM_002055.4(GFAP):c.232G>A (p.Asp78Asn)	GFAP	17	44915255	Alexander's disease	
NM_002055.4(GFAP):c.205G>A (p.Glu69Lys)	GFAP	17	44915282	Alexander's disease	
NM_002055.4(GFAP):c.197G>A (p.Arg66Gln)	GFAP	17	44915290	Alexander's disease	
NM_002055.4(GFAP):c.715C>T (p.Arg239Cys)	GFAP	17	44913334	Alexander's disease;not provided	
NM_002055.4(GFAP):c.716G>A (p.Arg239His)	GFAP	17	44913333	Alexander's disease;not provided	
NM_002055.4(GFAP):c.1246C>T (p.Arg416Trp)	GFAP	17	44908075	Alexander's disease;not provided	
NM_002055.4(GFAP):c.236G>A (p.Arg79His)	GFAP	17	44915251	Alexander's disease;not provided	
NM_002055.4(GFAP):c.235C>T (p.Arg79Cys)	GFAP	17	44915252	Alexander's disease;not provided	
NM_002055.4(GFAP):c.262C>T (p.Arg88Cys)	GFAP	17	44915225	Alexander's disease;not provided	
NM_002055.4(GFAP):c.226C>T (p.Leu76Phe)	GFAP	17	44915261	Alexander's disease;not provided	

NM_002055.4(GFAP):c.1091C>T (p.Ala364Val)	GFAP	17	44911272	Alexander's disease;not provided	
NM_002055.4(GFAP):c.1117G>A (p.Glu373Lys)	GFAP	17	44911246	Alexander's disease;not provided	
NM_002055.4(GFAP):c.1126C>T (p.Arg376Trp)	GFAP	17	44911237	Alexander's disease;not provided	
NM_002055.4(GFAP):c.1148C>T (p.Thr383Ile)	GFAP	17	44910638	Alexander's disease;not provided	
NM_002055.4(GFAP):c.208C>T (p.Arg70Trp)	GFAP	17	44915279	Alexander's disease;not provided	
NM_002055.4(GFAP):c.209G>A (p.Arg70Gln)	GFAP	17	44915278	Alexander's disease;not provided	
NM_002055.4(GFAP):c.214G>A (p.Glu72Lys)	GFAP	17	44915273	Alexander's disease;not provided	
NM_002055.4(GFAP):c.382G>A (p.Asp128Asn)	GFAP	17	44915105	Alexander's disease;not provided	
NM_002055.4(GFAP):c.613G>A (p.Glu205Lys)	GFAP	17	44913733	Alexander's disease;not provided	
NM_002055.4(GFAP):c.619G>A (p.Glu207Lys)	GFAP	17	44913430	Alexander's disease;not provided	
NM_002055.4(GFAP):c.628G>A (p.Glu210Lys)	GFAP	17	44913421	Alexander's disease;not provided	
NM_002055.4(GFAP):c.731C>T (p.Ala244Val)	GFAP	17	44913318	Alexander's disease;not provided	
NM_002055.4(GFAP):c.994G>A (p.Glu332Lys)	GFAP	17	44911369	Alexander's disease;not provided	
NM_001131019.2(GFAP):c.1289G>A (p.Arg430His)	GFAP	17	44910143	Alexander's disease;not provided	
NM_000187.3(HGD):c.688C>T (p.Pro230Ser)	HGD	3	120644405	Alkaptonuria	
NM_000187.3(HGD):c.481G>A (p.Gly161Arg)	HGD	3	120647041	Alkaptonuria	
NM_000187.3(HGD):c.16-1G>A	HGD	3	120675864	Alkaptonuria	
NM_000187.3(HGD):c.808G>A (p.Gly270Arg)	HGD	3	120641660	Alkaptonuria	
NM_000187.3(HGD):c.140C>T (p.Ser47Leu)	HGD	3	120674937	Alkaptonuria	
NM_000187.3(HGD):c.342+1G>A	HGD	3	120652591	Alkaptonuria	
NM_006517.4(SLC16A2):c.359C>T (p.Ser120Phe)	SLC16A2	X	74421996	Allan-Herndon-Dudley syndrome	
NM_006517.4(SLC16A2):c.277C>T (p.Gln93Ter)	SLC16A2	X	74421914	Allan-Herndon-Dudley syndrome	
NM_006517.4(SLC16A2):c.916C>T (p.Gln306Ter)	SLC16A2	X	74524699	Allan-Herndon-Dudley syndrome	
NM_006517.4(SLC16A2):c.326G>A (p.Trp109Ter)	SLC16A2	X	74421963	Allan-Herndon-Dudley syndrome	
NM_006517.4(SLC16A2):c.940C>T (p.Arg314Ter)	SLC16A2	X	74524723	Allan-Herndon-Dudley syndrome	
NM_006517.4(SLC16A2):c.-576-1G>A	SLC16A2	X	74524358	Allan-Herndon-Dudley syndrome	
NM_006517.4(SLC16A2):c.449C>T (p.Ala150Val)	SLC16A2	X	74521008	Allan-Herndon-Dudley syndrome;not provided	
NM_006517.4(SLC16A2):c.1111C>T (p.Arg371Cys)	SLC16A2	X	74525834	Allan-Herndon-Dudley syndrome;not provided	
NM_000477.5(ALB):c.71G>A (p.Arg24Gln)	ALB	4	73404398	Analbuminemia;Ehlers-Danlos syndrome, procollagen proteinase deficient	
NM_005144.4(HR):c.3034G>A (p.Asp1012Asn)	HR	8	22119227	Alopecia universalis congenita	
NM_024537.3(CARS2):c.752C>T (p.Pro251Leu)	CARS2	13	110677007	Alpers encephalopathy;Combined oxidative phosphorylation deficiency 27	
NM_000517.4(HBA2):c.69C>T (p.Gly23=)	HBA2	16	172981	Alpha plus thalassemia	
NM_000448.2(RAG1):c.2521C>T (p.Arg841Trp)	RAG1	11	36575825	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity	
NM_001127701.1(SERPINA1):c.1178C>T (p.Pro393Leu)	SERPINA1	14	94378528	Alpha-1-antitrypsin deficiency	
NM_001127701.1(SERPINA1):c.272G>A (p.Gly91Glu)	SERPINA1	14	94382966	Alpha-1-antitrypsin deficiency	
NM_001127701.1(SERPINA1):c.230C>T (p.Ser77Phe)	SERPINA1	14	94383008	Alpha-1-antitrypsin deficiency	
NM_000295.4(SERPINA1):c.538C>T (p.Gln180Ter)	SERPINA1	14	94382700	Alpha-1-antitrypsin deficiency	
NM_001127701.1(SERPINA1):c.1096G>A (p.Glu366Lys)	SERPINA1	14	94378610	Alpha-1-antitrypsin deficiency;FRAXE;not provided	
NM_000295.4(SERPINA1):c.1177C>T (p.Pro393Ser)	SERPINA1	14	94378529	Alpha-1-antitrypsin deficiency;not provided	
NM_001885.2(CRYAB):c.451C>T (p.Gln151Ter)	CRYAB	11	111908841	Alpha-B crystallinopathy	
NM_001134.2(AFP):c.543G>A (p.Trp181Ter)	AFP	4	73442356	Alpha-fetoprotein deficiency	
NM_000091.4(COL4A3):c.3499G>A (p.Gly1167Arg)	COL4A3	2	227295044	Alport syndrome, autosomal dominant	
NM_000091.4(COL4A3):c.4382C>T (p.Pro1461Leu)	COL4A3	2	227307839	Alport syndrome, autosomal dominant	
NM_000092.4(COL4A4):c.3601G>A (p.Gly1201Ser)	COL4A4	2	227032253	Alport syndrome, autosomal recessive	

NM_000092.4(COL4A4):c.4129C>T (p.Arg1377Ter)	COL4A4	2	227022135	Alport syndrome, autosomal recessive
NM_000092.4(COL4A4):c.4715C>T (p.Pro1572Leu)	COL4A4	2	227008112	Alport syndrome, autosomal recessive
NM_000091.4(COL4A3):c.4441C>T (p.Arg1481Ter)	COL4A3	2	227307898	Alport syndrome, autosomal recessive
NM_000091.4(COL4A3):c.3955G>A (p.Gly1319Arg)	COL4A3	2	227303110	Alport syndrome, autosomal recessive
NM_000495.4(COL4A5):c.3428G>A (p.Gly1143Asp)	COL4A5	X	108665561	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.974G>A (p.Gly325Glu)	COL4A5	X	108582921	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.161G>A (p.Gly54Asp)	COL4A5	X	108559083	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.5030G>A (p.Arg1677Gln)	COL4A5	X	108696350	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.142-1G>A	COL4A5	X	108559063	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.142G>A (p.Gly48Arg)	COL4A5	X	108559064	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.231+1G>A	COL4A5	X	108559154	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.232-1G>A	COL4A5	X	108563881	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.276+5G>A	COL4A5	X	108563931	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.286G>A (p.Gly96Arg)	COL4A5	X	108568638	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.322-1G>A	COL4A5	X	108568758	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.385-719G>A	COL4A5	X	108570694	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.385G>A (p.Gly129Arg)	COL4A5	X	108571413	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.386G>A (p.Gly129Glu)	COL4A5	X	108571414	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.430G>A (p.Gly144Ser)	COL4A5	X	108571458	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.431G>A (p.Gly144Asp)	COL4A5	X	108571459	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.438+5G>A	COL4A5	X	108571471	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.466-12G>A	COL4A5	X	108573562	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.538G>A (p.Gly180Arg)	COL4A5	X	108573646	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.546+1G>A	COL4A5	X	108573655	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.547-1G>A	COL4A5	X	108575909	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.574G>A (p.Gly192Arg)	COL4A5	X	108575937	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.584G>A (p.Gly195Asp)	COL4A5	X	108575947	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.593G>A (p.Gly198Glu)	COL4A5	X	108575956	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.609+1G>A	COL4A5	X	108575973	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.611G>A (p.Gly204Asp)	COL4A5	X	108577953	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.638G>A (p.Gly213Glu)	COL4A5	X	108577980	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.646G>A (p.Gly216Arg)	COL4A5	X	108578078	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.655G>A (p.Gly219Ser)	COL4A5	X	108578087	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.687+1G>A	COL4A5	X	108578120	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.689G>A (p.Gly230Asp)	COL4A5	X	108578292	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.716G>A (p.Gly239Glu)	COL4A5	X	108578319	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.791G>A (p.Gly264Asp)	COL4A5	X	108580543	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.796C>T (p.Arg266Ter)	COL4A5	X	108580548	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.834+1G>A	COL4A5	X	108580587	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.884G>A (p.Gly295Asp)	COL4A5	X	108580731	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.891+1G>A	COL4A5	X	108580739	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.892G>A (p.Gly298Ser)	COL4A5	X	108580983	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.920G>A (p.Gly307Asp)	COL4A5	X	108581011	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.928G>A (p.Gly310Arg)	COL4A5	X	108581019	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.937-1G>A	COL4A5	X	108582883	Alport syndrome, X-linked recessive

NM_000495.4(COL4A5):c.937G>A (p.Gly313Ser)	COL4A5	X	108582884	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.956G>A (p.Gly319Asp)	COL4A5	X	108582903	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.973G>A (p.Gly325Arg)	COL4A5	X	108582920	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1084G>A (p.Gly362Arg)	COL4A5	X	108586666	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1094G>A (p.Gly365Glu)	COL4A5	X	108586676	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1112G>A (p.Gly371Glu)	COL4A5	X	108586694	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1117C>T (p.Arg373Ter)	COL4A5	X	108586699	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1139G>A (p.Gly380Asp)	COL4A5	X	108586721	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1148G>A (p.Gly383Asp)	COL4A5	X	108586730	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1165+1G>A	COL4A5	X	108586748	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1199G>A (p.Gly400Glu)	COL4A5	X	108591091	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1219C>T (p.Gln407Ter)	COL4A5	X	108591111	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1226G>A (p.Gly409Asp)	COL4A5	X	108591118	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1243G>A (p.Gly415Arg)	COL4A5	X	108591135	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1259G>A (p.Gly420Glu)	COL4A5	X	108591151	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1268G>A (p.Gly423Glu)	COL4A5	X	108591160	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1276G>A (p.Gly426Arg)	COL4A5	X	108591168	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1286G>A (p.Gly429Glu)	COL4A5	X	108591178	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1294G>A (p.Gly432Arg)	COL4A5	X	108591186	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1397G>A (p.Gly466Glu)	COL4A5	X	108591618	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1406G>A (p.Gly469Glu)	COL4A5	X	108591627	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1414G>A (p.Gly472Arg)	COL4A5	X	108591635	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1423G>A (p.Gly475Ser)	COL4A5	X	108591644	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1423+1G>A	COL4A5	X	108591645	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1424-1G>A	COL4A5	X	108595508	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1472G>A (p.Gly491Glu)	COL4A5	X	108595557	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1481G>A (p.Gly494Asp)	COL4A5	X	108595566	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1516+1G>A	COL4A5	X	108595602	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1562G>A (p.Gly521Asp)	COL4A5	X	108597043	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1571G>A (p.Gly524Asp)	COL4A5	X	108597052	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1587+1G>A	COL4A5	X	108597069	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1598G>A (p.Gly533Glu)	COL4A5	X	108597387	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1607G>A (p.Gly536Asp)	COL4A5	X	108597396	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1643G>A (p.Gly548Asp)	COL4A5	X	108597432	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1681G>A (p.Gly561Arg)	COL4A5	X	108597470	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1718G>A (p.Gly573Asp)	COL4A5	X	108597507	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1726G>A (p.Gly576Ser)	COL4A5	X	108597515	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1735G>A (p.Gly579Arg)	COL4A5	X	108597524	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1736G>A (p.Gly579Glu)	COL4A5	X	108597525	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1738C>T (p.Gln580Ter)	COL4A5	X	108597527	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1780-1G>A	COL4A5	X	108598701	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1780G>A (p.Gly594Ser)	COL4A5	X	108598702	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1783G>A (p.Gly595Arg)	COL4A5	X	108598705	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1856C>T (p.Pro619Leu)	COL4A5	X	108598778	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1886G>A (p.Gly629Asp)	COL4A5	X	108598808	Alport syndrome, X-linked recessive

NM_000495.4(COL4A5):c.1895G>A (p.Gly632Asp)	COL4A5	X	108598817	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1897G>A (p.Glu633Lys)	COL4A5	X	108598819	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1904G>A (p.Gly635Asp)	COL4A5	X	108598826	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1912G>A (p.Gly638Ser)	COL4A5	X	108598834	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1948+1G>A	COL4A5	X	108598871	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1957G>A (p.Gly653Arg)	COL4A5	X	108601401	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.1997G>A (p.Gly666Asp)	COL4A5	X	108601441	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2023G>A (p.Gly675Ser)	COL4A5	X	108601467	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2042G>A (p.Gly681Asp)	COL4A5	X	108601885	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2060G>A (p.Gly687Glu)	COL4A5	X	108601903	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2165G>A (p.Gly722Glu)	COL4A5	X	108602982	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2215C>T (p.Pro739Ser)	COL4A5	X	108603032	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2219G>A (p.Gly740Glu)	COL4A5	X	108603036	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2228G>A (p.Gly743Asp)	COL4A5	X	108603045	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2245-1G>A	COL4A5	X	108606741	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2287G>A (p.Gly763Arg)	COL4A5	X	108606784	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2288G>A (p.Gly763Glu)	COL4A5	X	108606785	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2297G>A (p.Gly766Asp)	COL4A5	X	108606794	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2305G>A (p.Gly769Arg)	COL4A5	X	108606802	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2315G>A (p.Gly772Asp)	COL4A5	X	108606812	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2332G>A (p.Gly778Ser)	COL4A5	X	108606829	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2386G>A (p.Gly796Arg)	COL4A5	X	108606883	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2404G>A (p.Gly802Arg)	COL4A5	X	108614919	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2423G>A (p.Gly808Glu)	COL4A5	X	108614938	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2431G>A (p.Gly811Arg)	COL4A5	X	108614946	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2509G>A	COL4A5	X	108615024	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2554G>A (p.Gly852Arg)	COL4A5	X	108620303	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2555G>A (p.Gly852Glu)	COL4A5	X	108620304	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2597G>A (p.Gly866Glu)	COL4A5	X	108620346	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2624G>A (p.Gly875Glu)	COL4A5	X	108620373	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2705G>A (p.Gly902Glu)	COL4A5	X	108621830	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2722G>A (p.Gly908Arg)	COL4A5	X	108621847	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2731G>A (p.Gly911Arg)	COL4A5	X	108621856	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2732G>A (p.Gly911Glu)	COL4A5	X	108621857	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2782C>T (p.Gln928Ter)	COL4A5	X	108622690	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2788C>T (p.Gln930Ter)	COL4A5	X	108622696	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2804G>A (p.Gly935Asp)	COL4A5	X	108622712	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.2840G>A (p.Gly947Asp)	COL4A5	X	108622748	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3017-1G>A	COL4A5	X	108625704	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3046C>T (p.Gln1016Ter)	COL4A5	X	108625734	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3088G>A (p.Gly1030Ser)	COL4A5	X	108625776	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3115G>A (p.Gly1039Ser)	COL4A5	X	108626218	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3134G>A (p.Gly1045Glu)	COL4A5	X	108626237	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3181C>T (p.Gln1061Ter)	COL4A5	X	108626284	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3196G>A (p.Gly1066Ser)	COL4A5	X	108626299	Alport syndrome, X-linked recessive

NM_000495.4(COL4A5):c.3247G>A (p.Gly1083Ser)	COL4A5	X	108655331	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3257G>A (p.Gly1086Asp)	COL4A5	X	108655341	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3319G>A (p.Gly1107Arg)	COL4A5	X	108655403	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3373+1G>A	COL4A5	X	108655458	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3427G>A (p.Gly1143Ser)	COL4A5	X	108665560	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3481G>A (p.Gly1161Arg)	COL4A5	X	108666522	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3499G>A (p.Gly1167Ser)	COL4A5	X	108666540	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3508G>A (p.Gly1170Ser)	COL4A5	X	108666549	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3535G>A (p.Gly1179Arg)	COL4A5	X	108666576	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3538C>T (p.Gln1180Ter)	COL4A5	X	108666579	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3554-1G>A	COL4A5	X	108667132	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3586G>A (p.Gly1196Arg)	COL4A5	X	108667165	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3587G>A (p.Gly1196Glu)	COL4A5	X	108667166	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3605-1G>A	COL4A5	X	108668318	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3613G>A (p.Gly1205Ser)	COL4A5	X	108668327	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3632G>A (p.Gly1211Glu)	COL4A5	X	108668346	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3641G>A (p.Gly1214Glu)	COL4A5	X	108668355	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3659G>A (p.Gly1220Asp)	COL4A5	X	108668373	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3686G>A (p.Gly1229Asp)	COL4A5	X	108668400	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3694G>A (p.Gly1232Ser)	COL4A5	X	108668408	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3700C>T (p.Gln1234Ter)	COL4A5	X	108668414	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3731G>A (p.Gly1244Asp)	COL4A5	X	108668445	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3754G>A (p.Gly1252Ser)	COL4A5	X	108668468	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3763G>A (p.Gly1255Arg)	COL4A5	X	108668477	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3782G>A (p.Gly1261Glu)	COL4A5	X	108668496	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3808G>A (p.Gly1270Ser)	COL4A5	X	108677517	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3925-1G>A	COL4A5	X	108680678	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.3997G>A (p.Gly1333Ser)	COL4A5	X	108680751	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4069G>A (p.Gly1357Ser)	COL4A5	X	108680956	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4069+1G>A	COL4A5	X	108680957	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4147C>T (p.Gln1383Ter)	COL4A5	X	108681837	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4177C>T (p.Gln1393Ter)	COL4A5	X	108681867	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4199-1G>A	COL4A5	X	108686030	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4228C>T (p.Arg1410Cys)	COL4A5	X	108686060	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4297+1G>A	COL4A5	X	108686130	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4351G>A (p.Gly1451Ser)	COL4A5	X	108687535	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4325G>A (p.Gly1442Asp)	COL4A5	X	108687509	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4342G>A (p.Gly1448Ser)	COL4A5	X	108687526	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4463C>T (p.Ser1488Phe)	COL4A5	X	108687647	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4501C>T (p.Gln1501Ter)	COL4A5	X	108687685	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4614G>A (p.Trp1538Ter)	COL4A5	X	108692851	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4688G>A (p.Arg1563Gln)	COL4A5	X	108692925	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4702G>A (p.Glu1568Lys)	COL4A5	X	108694820	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4751C>T (p.Pro1584Leu)	COL4A5	X	108694869	Alport syndrome, X-linked recessive
NM_000495.4(COL4A5):c.4787G>A (p.Gly1596Asp)	COL4A5	X	108694905	Alport syndrome, X-linked recessive

NM_000495.4(COL4A5):c.4803G>A (p.Met1601Ile)	COL4A5	X	108694921	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.4803+1G>A	COL4A5	X	108694922	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.4913G>A (p.Cys1638Tyr)	COL4A5	X	108695376	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.4976G>A (p.Ser1659Asn)	COL4A5	X	108695439	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.5020C>T (p.Arg1674Ter)	COL4A5	X	108696340	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.5029C>T (p.Arg1677Ter)	COL4A5	X	108696349	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.1135C>T (p.Gln379Ter)	COL4A5	X	108586717	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.1216G>A (p.Gly406Ser)	COL4A5	X	108591108	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.1589G>A (p.Gly530Asp)	COL4A5	X	108597378	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.1744G>A (p.Gly582Arg)	COL4A5	X	108597533	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.1835G>A (p.Gly612Asp)	COL4A5	X	108598757	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.1883C>T (p.Pro628Leu)	COL4A5	X	108598805	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.2483G>A (p.Gly828Glu)	COL4A5	X	108614998	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.4271G>A (p.Gly1424Glu)	COL4A5	X	108686103	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.1844G>A (p.Gly615Glu)	COL4A5	X	108598766	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.3998-1G>A	COL4A5	X	108680884	Alport syndrome, X-linked recessive	
NM_033380.2(COL4A5):c.2395+1G>A	COL4A5	X	108606893	Alport syndrome, X-linked recessive	
NM_033380.2(COL4A5):c.2741G>A (p.Gly914Asp)	COL4A5	X	108621866	Alport syndrome, X-linked recessive	
NM_000495.4(COL4A5):c.1871G>A (p.Gly624Asp)	COL4A5	X	108598793	Alport syndrome, X-linked recessive;Glomerulopathy;Hypertension;Mild proteinuria;not provided	
NM_000495.4(COL4A5):c.2605G>A (p.Gly869Arg)	COL4A5	X	108620354	Alport syndrome, X-linked recessive;not provided	
NM_000495.4(COL4A5):c.4687C>T (p.Arg1563Ter)	COL4A5	X	108692924	Alport syndrome, X-linked recessive;not provided	
NM_014026.5(DCPS):c.636+1G>A	DCPS	11	126338400	AL-RAQAD SYNDROME	
NM_014026.5(DCPS):c.947C>T (p.Thr316Met)	DCPS	11	126345546	AL-RAQAD SYNDROME	
NM_015120.4(ALMS1):c.8383C>T (p.Gln2795Ter)	ALMS1	2	73490339	Alstrom syndrome	
NM_015120.4(ALMS1):c.10992G>A (p.Trp3664Ter)	ALMS1	2	73572866	Alstrom syndrome	
NM_015120.4(ALMS1):c.8164C>T (p.Arg2722Ter)	ALMS1	2	73490120	Alstrom syndrome	
NM_015120.4(ALMS1):c.6436C>T (p.Arg2146Ter)	ALMS1	2	73452960	Alstrom syndrome	
NM_015120.4(ALMS1):c.10483C>T (p.Gln3495Ter)	ALMS1	2	73572357	Alstrom syndrome	
NM_015120.4(ALMS1):c.5590C>T (p.Gln1864Ter)	ALMS1	2	73452114	Alstrom syndrome	
NM_015120.4(ALMS1):c.1900C>T (p.Gln634Ter)	ALMS1	2	73448424	Alstrom syndrome;not provided	
NM_152296.4(ATP1A3):c.2780G>A (p.Cys927Tyr)	ATP1A3	19	41968824	Alternating hemiplegia of childhood 2	
NM_152296.4(ATP1A3):c.2542+1G>A	ATP1A3	19	41970184	Alternating hemiplegia of childhood 2	
NM_152296.4(ATP1A3):c.2411C>T (p.Thr804Ile)	ATP1A3	19	41970395	Alternating hemiplegia of childhood 2	
NM_152296.4(ATP1A3):c.2263G>A (p.Gly755Ser)	ATP1A3	19	41975629	Alternating hemiplegia of childhood 2	
NM_152296.4(ATP1A3):c.2452G>A (p.Glu818Lys)	ATP1A3	19	41970275	Alternating hemiplegia of childhood 2;Cerebellar ataxia, areflexia, pes cavus, optic atrophy and sensorinural hearing loss;Dystonia 12;Inborn genetic diseases;not provided	
NM_152296.4(ATP1A3):c.2767G>A (p.Asp923Asn)	ATP1A3	19	41968837	Alternating hemiplegia of childhood 2;Dystonia 12	
NM_152296.4(ATP1A3):c.2600G>A (p.Gly867Asp)	ATP1A3	19	41969523	Alternating hemiplegia of childhood 2;Dystonia 12	
NM_152296.4(ATP1A3):c.2839G>A (p.Gly947Arg)	ATP1A3	19	41967744	Alternating hemiplegia of childhood 2;Dystonia 12;Hemiplegia;Seizure Disorders;not provided	
NM_152296.4(ATP1A3):c.2443G>A (p.Glu815Lys)	ATP1A3	19	41970284	Alternating hemiplegia of childhood 2;Dystonia 12;not provided	
NM_152296.4(ATP1A3):c.2401G>A (p.Asp801Asn)	ATP1A3	19	41970405	Alternating hemiplegia of childhood 2;not provided	
NM_000021.3(PSEN1):c.1141C>T (p.Leu381Phe)	PSEN1	14	73217137	Alzheimer disease familial 3, with spastic paraparesis	
NM_000021.3(PSEN1):c.796G>A (p.Gly266Ser)	PSEN1	14	73198057	Alzheimer disease, familial, 3, with spastic paraparesis and apraxia	
NM_000484.3(APP):c.2017G>A (p.Ala673Thr)	APP	21	25897620	Alzheimer disease, protection against;not provided	
NM_000484.3(APP):c.2149G>A (p.Val717Ile)	APP	21	25891784	Alzheimer disease, type 1;Alzheimer's disease;not provided	

NM_000484.3(APP):c.2137G>A (p.Ala713Thr)	APP	21	25891796	Alzheimer disease, type 1;not provided
NM_000484.3(APP):c.2143G>A (p.Val715Met)	APP	21	25891790	Alzheimer disease, type 1;not provided
NM_000484.3(APP):c.2141C>T (p.Thr714Ile)	APP	21	25891792	Alzheimer disease, type 1;not provided
NM_000021.3(PSEN1):c.438G>A (p.Met146Ile)	PSEN1	14	73173665	Alzheimer disease, type 3
NM_000021.3(PSEN1):c.1229G>A (p.Cys410Tyr)	PSEN1	14	73217225	Alzheimer disease, type 3;not provided
NM_000021.3(PSEN1):c.487C>T (p.His163Tyr)	PSEN1	14	73186859	Alzheimer disease, type 3;not provided
NM_000021.3(PSEN1):c.799C>T (p.Pro267Ser)	PSEN1	14	73198060	Alzheimer disease, type 3;not provided
NM_000021.3(PSEN1):c.236C>T (p.Ala79Val)	PSEN1	14	73170945	Alzheimer disease, type 3;not provided
NM_000021.3(PSEN1):c.509C>T (p.Ser170Phe)	PSEN1	14	73186881	Alzheimer disease, type 3;not provided
NM_000447.2(PSEN2):c.1289C>T (p.Thr430Met)	PSEN2	1	226895521	Alzheimer disease, type 4;not provided
NM_000447.2(PSEN2):c.717G>A (p.Met239Ile)	PSEN2	1	226888979	Alzheimer disease, type 4;not provided
NM_000447.2(PSEN2):c.254C>T (p.Ala85Val)	PSEN2	1	226883817	Alzheimer disease, type 4;not provided
NM_000021.3(PSEN1):c.806G>A (p.Arg269His)	PSEN1	14	73198067	Alzheimer disease, type 4;not provided
NM_000484.3(APP):c.2018C>T (p.Ala673Val)	APP	21	25897619	Alzheimer's disease
NM_031889.2(ENAM):c.534+1G>A	ENAM	4	70635895	Amelogenesis imperfecta - hypoplastic autosomal dominant - local
NM_198488.3(FAM83H):c.973C>T (p.Arg325Ter)	FAM83H	8	143728488	Amelogenesis imperfecta, hypocalcification type
NM_198488.3(FAM83H):c.1192C>T (p.Gln398Ter)	FAM83H	8	143728269	Amelogenesis imperfecta, hypocalcification type
NM_198488.3(FAM83H):c.1380G>A (p.Trp460Ter)	FAM83H	8	143728081	Amelogenesis imperfecta, hypocalcification type
NM_198488.3(FAM83H):c.2029C>T (p.Gln677Ter)	FAM83H	8	143727432	Amelogenesis imperfecta, hypocalcification type
NM_198488.3(FAM83H):c.1408C>T (p.Gln470Ter)	FAM83H	8	143728053	Amelogenesis imperfecta, hypocalcification type
NM_198488.3(FAM83H):c.1379G>A (p.Trp460Ter)	FAM83H	8	143728082	Amelogenesis imperfecta, hypocalcification type
NM_198488.3(FAM83H):c.1366C>T (p.Gln456Ter)	FAM83H	8	143728095	Amelogenesis imperfecta, hypocalcification type
NM_178497.3(C4orf26):c.318G>A	C4orf26	4	75564364	Amelogenesis imperfecta, hypomaturation type IIA4
NM_153648.3(SLC24A4):c.823C>T (p.Arg275Ter)	SLC24A4	14	92454034	Amelogenesis imperfecta, hypomaturation type IIA5
NM_153648.3(SLC24A4):c.245C>T (p.Ala82Val)	SLC24A4	14	92442132	Amelogenesis imperfecta, hypomaturation type IIA5
NM_004917.4(KLK4):c.458G>A (p.Trp153Ter)	KLK4	19	50908596	Amelogenesis imperfecta, hypomaturation type, IIA1
NM_004771.3(MMP20):c.102G>A (p.Trp34Ter)	MMP20	11	102625218	Amelogenesis imperfecta, hypomaturation type, IIA2
NM_182758.3(WDR72):c.2934G>A (p.Trp978Ter)	WDR72	15	53609531	Amelogenesis imperfecta, hypomaturation type, IIA3;not provided
NM_182680.1(AMELX):c.152C>T (p.Thr51Ile)	AMELX	X	11298243	Amelogenesis imperfecta, type 1E
NM_000888.4(ITGB6):c.427G>A (p.Ala143Thr)	ITGB6	2	160195535	Amelogenesis imperfecta, type IH
NM_000888.4(ITGB6):c.1846C>T (p.Arg616Ter)	ITGB6	2	160126416	Amelogenesis imperfecta, type IH
NM_033068.2(ACPT):c.713C>T (p.Ser238Leu)	ACPT	19	50793751	AMELOGENESIS IMPERFECTA, TYPE IJ
NM_033068.2(ACPT):c.331C>T (p.Arg111Cys)	ACPT	19	50791683	AMELOGENESIS IMPERFECTA, TYPE IJ
NM_033068.2(ACPT):c.226C>T (p.Arg76Cys)	ACPT	19	50790783	AMELOGENESIS IMPERFECTA, TYPE IJ
NM_033068.2(ACPT):c.397G>A (p.Glu133Lys)	ACPT	19	50791749	AMELOGENESIS IMPERFECTA, TYPE IJ
NM_000666.2(ACY1):c.360-1G>A	ACY1	3	51986254	Aminoacylase 1 deficiency
NM_000666.2(ACY1):c.589C>T (p.Arg197Trp)	ACY1	3	51986993	Aminoacylase 1 deficiency
m.7444G>A	MT-CO1;MT-	MT	7444	Aminoglycoside-induced deafness;Deafness, nonsyndromic sensorineural, mitochondrial;Leber's optic atrophy
NM_003896.3(ST3GAL5):c.862C>T (p.Arg288Ter)	ST3GAL5	2	85844542	Amish infantile epilepsy syndrome
NM_003896.3(ST3GAL5):c.1063G>A (p.Glu355Lys)	ST3GAL5	2	85840338	Amish infantile epilepsy syndrome
NM_003896.3(ST3GAL5):c.601G>A (p.Gly201Arg)	ST3GAL5	2	85847922	Amish infantile epilepsy syndrome
NM_000371.3(TTR):c.424G>A (p.Val142Ile)	TTR	18	31598655	Amyloid Cardiomyopathy, Transthyretin-related;Amyloidogenic transthyretin amyloidosis;Cardiomyopathy;Cardiovascular phenotype;not provided
NM_000371.3(TTR):c.133G>A (p.Ala45Thr)	TTR	18	31592959	Amyloidogenic transthyretin amyloidosis

NM_000371.3(TTR):c.241G>A (p.Glu81Lys)	TTR	18	31595160	Amyloidogenic transthyretin amyloidosis
NM_000371.3(TTR):c.199G>A (p.Gly67Arg)	TTR	18	31593025	Amyloidogenic transthyretin amyloidosis
NM_000371.3(TTR):c.148G>A (p.Val50Met)	TTR	18	31592974	Amyloidogenic transthyretin amyloidosis;not provided
NM_000371.3(TTR):c.118G>A (p.Val40Ile)	TTR	18	31592944	Amyloidogenic transthyretin amyloidosis;not provided
NM_000371.3(TTR):c.218G>A (p.Gly73Glu)	TTR	18	31595137	AMYLOIDOSIS, LEPTOMENINGEAL, TRANSTHYRETIN-RELATED
NM_000454.4(SOD1):c.289G>A (p.Asp97Asn)	SOD1	21	31667307	Amyotrophic lateral sclerosis 1, autosomal recessive
NM_007126.3(VCP):c.1774G>A (p.Asp592Asn)	VCP	9	35059723	Amyotrophic lateral sclerosis 14 without frontotemporal dementia
NM_007126.3(VCP):c.464G>A (p.Arg155His)	VCP	9	35065363	Amyotrophic lateral sclerosis 14 without frontotemporal dementia;Inclusion body myopathy with early-onset paget disease and frontotemporal dementia
NM_007126.3(VCP):c.572G>A (p.Arg191Gln)	VCP	9	35065255	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia;Inclusion body myopathy with early-onset paget disease and frontotemporal dementia
NM_013444.3(UBQLN2):c.1489C>T (p.Pro497Ser)	UBQLN2	X	56565362	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia
NM_013444.3(UBQLN2):c.1525C>T (p.Pro509Ser)	UBQLN2	X	56565398	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia
NM_013444.3(UBQLN2):c.1573C>T (p.Pro525Ser)	UBQLN2	X	56565446	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia
NM_013444.3(UBQLN2):c.1490C>T (p.Pro497Leu)	UBQLN2	X	56565363	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia
NM_005235.2(ERBB4):c.2780G>A (p.Arg927Gln)	ERBB4	2	211424241	Amyotrophic lateral sclerosis 19;not provided
NM_005235.2(ERBB4):c.3823C>T (p.Arg1275Trp)	ERBB4	2	211383719	Amyotrophic lateral sclerosis 19;not provided
NM_031157.3(HNRNPA1):c.940G>A (p.Asp314Asn)	HNRNPA1	12	54283844	Amyotrophic lateral sclerosis 20
NM_006000.2(TUBA4A):c.643C>T (p.Arg215Cys)	TUBA4A	2	219251056	Amyotrophic lateral sclerosis 22 with frontotemporal dementia
NM_006000.2(TUBA4A):c.958C>T (p.Arg320Cys)	TUBA4A	2	219250741	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia
NM_006000.2(TUBA4A):c.959G>A (p.Arg320His)	TUBA4A	2	219250740	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia
NM_006000.2(TUBA4A):c.1220G>A (p.Trp407Ter)	TUBA4A	2	219250479	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia
NM_006000.2(TUBA4A):c.1147G>A (p.Ala383Thr)	TUBA4A	2	219250552	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia
NM_000454.4(SOD1):c.112G>A (p.Gly38Arg)	SOD1	21	31663829	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.124G>A (p.Gly42Ser)	SOD1	21	31663841	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.125G>A (p.Gly42Asp)	SOD1	21	31663842	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.14C>T (p.Ala5Val)	SOD1	21	31659783	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.13G>A (p.Ala5Thr)	SOD1	21	31659782	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.436G>A (p.Ala146Thr)	SOD1	21	31668549	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.64G>A (p.Glu22Lys)	SOD1	21	31659833	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.404G>A (p.Ser135Asn)	SOD1	21	31668517	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.49G>A (p.Gly17Ser)	SOD1	21	31659818	Amyotrophic lateral sclerosis type 1
NM_000454.4(SOD1):c.217G>A (p.Gly73Ser)	SOD1	21	31666496	Amyotrophic lateral sclerosis type 1
NM_007375.3(TARDBP):c.892G>A (p.Gly298Ser)	TARDBP	1	11022301	Amyotrophic lateral sclerosis type 10
NM_007375.3(TARDBP):c.943G>A (p.Ala315Thr)	TARDBP	1	11022352	Amyotrophic lateral sclerosis type 10
NM_007375.3(TARDBP):c.883G>A (p.Gly295Ser)	TARDBP	1	11022292	Amyotrophic lateral sclerosis type 10
NM_007375.3(TARDBP):c.*697G>A	TARDBP	1	11023351	Amyotrophic lateral sclerosis type 10;FRONTOTEMPORAL DEMENTIA WITH TDP43 INCLUSIONS, TARDBP-RELATED
NM_007375.3(TARDBP):c.1144G>A (p.Ala382Thr)	TARDBP	1	11022553	Amyotrophic lateral sclerosis type 10;FRONTOTEMPORAL DEMENTIA WITH TDP43 INCLUSIONS, TARDBP-RELATED;not provided
NM_007375.3(TARDBP):c.859G>A (p.Gly287Ser)	TARDBP	1	11022268	Amyotrophic lateral sclerosis type 10;not provided
NM_014845.5(FIG4):c.547C>T (p.Arg183Ter)	FIG4	6	109735199	Amyotrophic lateral sclerosis type 11;Charcot-Marie-Tooth disease, type 4J;not provided
NM_001008211.1(OPTN):c.1192C>T (p.Gln398Ter)	OPTN	10	13125989	Amyotrophic lateral sclerosis type 12
NM_015046.5(SETX):c.6407G>A (p.Arg2136His)	SETX	9	132283403	Amyotrophic lateral sclerosis type 4
NM_015046.5(SETX):c.8C>T (p.Thr3Ile)	SETX	9	132349421	Amyotrophic lateral sclerosis type 4;not provided

NM_025137.3(SPG11):c.118C>T (p.Gln40Ter)	SPG11	15	44663530	Amyotrophic lateral sclerosis type 5;Charcot-Marie-Tooth disease, axonal type 2X;Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.267G>A (p.Trp89Ter)	SPG11	15	44660607	Amyotrophic lateral sclerosis type 5;Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.5974C>T (p.Arg1992Ter)	SPG11	15	44574934	Amyotrophic lateral sclerosis type 5;Spastic paraplegia 11, autosomal recessive;not provided
NM_004960.3(FUS):c.1553G>A (p.Arg518Lys)	FUS	16	31191410	Amyotrophic lateral sclerosis type 6
NM_004960.3(FUS):c.1561C>T (p.Arg521Cys)	FUS	16	31191418	Amyotrophic lateral sclerosis type 6
NM_004960.3(FUS):c.1562G>A (p.Arg521His)	FUS	16	31191419	Amyotrophic lateral sclerosis type 6
NM_004960.3(FUS):c.1520G>A (p.Gly507Asp)	FUS	16	31191089	Amyotrophic lateral sclerosis type 6
NM_004960.3(FUS):c.1483C>T (p.Arg495Ter)	FUS	16	31191052	Amyotrophic lateral sclerosis type 6
NM_004960.3(FUS):c.616G>A (p.Gly206Ser)	FUS	16	31185031	Amyotrophic lateral sclerosis type 6
NM_004960.3(FUS):c.646C>T (p.Arg216Cys)	FUS	16	31185061	Amyotrophic lateral sclerosis type 6;Tremor, hereditary essential, 4
NM_004738.4(VAPB):c.166C>T (p.Pro56Ser)	VAPB	20	58418318	Amyotrophic lateral sclerosis type 8;Amyotrophic lateral sclerosis, typical;Spinal muscular atrophy, late-onset, finkel type;not provided
NM_004738.4(VAPB):c.137C>T (p.Thr46Ile)	VAPB	20	58418289	Amyotrophic lateral sclerosis type 8;not provided
NM_001145.4(ANG):c.164G>A (p.Arg55Lys)	ANG	14	20693728	Amyotrophic lateral sclerosis type 9
NM_001145.4(ANG):c.155G>A (p.Ser52Asn)	ANG	14	20693719	Amyotrophic lateral sclerosis type 9
NM_001145.4(ANG):c.407C>T (p.Pro136Leu)	ANG	14	20693971	Amyotrophic lateral sclerosis type 9
NM_001145.4(ANG):c.409G>A (p.Val137Ile)	ANG	14	20693973	Amyotrophic lateral sclerosis type 9
NM_006640.4(SEPT9):c.262C>T (p.Arg88Trp)	9-Sep	17	77402298	Amyotrophy, hereditary neuralgic
NM_006640.4(SEPT9):c.278C>T (p.Ser93Phe)	9-Sep	17	77402314	Amyotrophy, hereditary neuralgic
NM_002296.3(LBR):c.226C>T (p.Arg76Ter)	LBR	1	225422217	Anadysplasia-like, spontaneously remitting spondylometaphyseal dysplasia
NM_000477.6(ALB):c.79+1G>A	ALB	4	73404407	Analbuminemia baghdad
NM_001145860.1(POP1):c.1573C>T (p.Pro525Ser)	POP1	8	98140867	ANAUXETIC DYSPLASIA 2
NM_001145860.1(POP1):c.1748G>A (p.Gly583Glu)	POP1	8	98148852	ANAUXETIC DYSPLASIA 2
NM_001145860.1(POP1):c.1744C>T (p.Pro582Ser)	POP1	8	98148848	ANAUXETIC DYSPLASIA 2
NM_133647.1(SLC12A6):c.3031C>T (p.Arg1011Ter)	SLC12A6	15	34236719	Andermann syndrome
NM_133647.1(SLC12A6):c.2023C>T (p.Arg675Ter)	SLC12A6	15	34243993	Andermann syndrome
NM_005135.2(SLC12A6):c.466C>T (p.Arg156Cys)	SLC12A6	15	34257713	Andermann syndrome
NM_005135.2(SLC12A6):c.3247C>T (p.Arg1083Ter)	SLC12A6	15	34233934	Andermann syndrome
NM_000891.2(KCNJ2):c.557C>T (p.Pro186Leu)	KCNJ2	17	70175596	Andersen Tawil syndrome;Congenital long QT syndrome
NM_000891.2(KCNJ2):c.904G>A (p.Val302Met)	KCNJ2	17	70175943	Andersen Tawil syndrome;Congenital long QT syndrome
NM_000891.2(KCNJ2):c.245G>A (p.Arg82Gln)	KCNJ2	17	70175284	Andersen Tawil syndrome;Congenital long QT syndrome
NM_000891.2(KCNJ2):c.644G>A (p.Gly215Asp)	KCNJ2	17	70175683	Andersen Tawil syndrome;Congenital long QT syndrome
NM_000891.2(KCNJ2):c.899G>A (p.Gly300Asp)	KCNJ2	17	70175938	Andersen Tawil syndrome;Congenital long QT syndrome
NM_000891.2(KCNJ2):c.652C>T (p.Arg218Trp)	KCNJ2	17	70175691	Andersen Tawil syndrome;Congenital long QT syndrome;not provided
NM_000891.2(KCNJ2):c.199C>T (p.Arg67Trp)	KCNJ2	17	70175238	Andersen Tawil syndrome;Congenital long QT syndrome;not provided
NM_000891.2(KCNJ2):c.653G>A (p.Arg218Gln)	KCNJ2	17	70175692	Andersen Tawil syndrome;Congenital long QT syndrome;not provided
NM_000044.4(AR):c.1826G>A (p.Arg609Lys)	AR	X	67686067	Androgen insensitivity, partial, with breast cancer
NM_000044.4(AR):c.2323C>T (p.Arg775Cys)	AR	X	67721837	Androgen resistance syndrome
NM_000044.4(AR):c.2157G>A (p.Trp719Ter)	AR	X	67711673	Androgen resistance syndrome
NM_000044.4(AR):c.2599G>A (p.Val867Met)	AR	X	67722976	Androgen resistance syndrome
NM_000044.4(AR):c.2391G>A (p.Trp797Ter)	AR	X	67721905	Androgen resistance syndrome
NM_000044.4(AR):c.2324G>A (p.Arg775His)	AR	X	67721838	Androgen resistance syndrome
NM_000044.4(AR):c.178C>T (p.Gln60Ter)	AR	X	67545324	Androgen resistance syndrome
NM_000044.4(AR):c.2667C>T (p.Ser889=)	AR	X	67723745	Androgen resistance syndrome

NM_000044.4(AR):c.2137C>T (p.Leu713Phe)	AR	X	67711653	Androgen resistance syndrome
NM_000044.4(AR):c.1732G>A (p.Gly578Arg)	AR	X	67643371	Androgen resistance syndrome
NM_000044.4(AR):c.2231G>A (p.Gly744Glu)	AR	X	67717535	Androgen resistance syndrome
NM_000044.4(AR):c.2521C>T (p.Arg841Cys)	AR	X	67722898	Androgen resistance syndrome;Reifenstein syndrome
NM_004299.4(ABCB7):c.1300G>A (p.Glu434Lys)	ABCB7	X	75070433	Anemia sideroblastic and spinocerebellar ataxia
NM_000402.4(G6PD):c.1172C>T (p.Ala391Val)	G6PD	X	154532772	Anemia, nonspeherocytic hemolytic, due to g6pd deficiency
NM_000402.4(G6PD):c.577G>A (p.Gly193Ser)	G6PD	X	154534495	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
NM_000402.4(G6PD):c.1268G>A (p.Arg423His)	G6PD	X	154532676	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
NM_000402.4(G6PD):c.1192G>A (p.Glu398Lys)	G6PD	X	154532752	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
NM_000402.4(G6PD):c.1249C>T (p.Arg417Cys)	G6PD	X	154532695	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
NM_000402.4(G6PD):c.1319G>A (p.Gly440Asp)	G6PD	X	154532625	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
NM_000402.4(G6PD):c.896G>A (p.Cys299Tyr)	G6PD	X	154533634	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
NM_000402.4(G6PD):c.653C>T (p.Ser218Phe)	G6PD	X	154534419	Anemia, nonspherocytic hemolytic, due to G6PD deficiency;Favism, susceptibility to;Glucose 6 phosphate dehydrogenase deficiency;Susceptibility to malaria;not provided
NM_000402.4(G6PD):c.1450C>T (p.Arg484Cys)	G6PD	X	154532390	Anemia, nonspherocytic hemolytic, due to G6PD deficiency;Favism, susceptibility to;not provided
NM_000402.4(G6PD):c.961G>A (p.Val321Met)	G6PD	X	154533122	Anemia, nonspherocytic hemolytic, due to G6PD deficiency;Glucose 6 phosphate dehydrogenase deficiency;not provided
NM_000402.4(G6PD):c.1093G>A (p.Ala365Thr)	G6PD	X	154532990	Anemia, nonspherocytic hemolytic, due to G6PD deficiency;not provided
NM_000402.4(G6PD):c.1429G>A (p.Gly477Arg)	G6PD	X	154532411	Anemia, nonspherocytic hemolytic, due to G6PD deficiency;not provided
NM_000402.4(G6PD):c.1478G>A (p.Arg493His)	G6PD	X	154532257	Anemia, nonspherocytic hemolytic, due to G6PD deficiency;not provided
NM_017875.2(SLC25A38):c.349C>T (p.Arg117Ter)	SLC25A38	3	39391513	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive
NM_017875.2(SLC25A38):c.277-1G>A	SLC25A38	3	39391440	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive
NM_130838.1(UBE3A):c.1249C>T (p.Arg417Ter)	UBE3A	15	25370865	Angelman syndrome
NM_130838.1(UBE3A):c.2304G>A (p.Trp768Ter)	UBE3A	15	25340219	Angelman syndrome
NM_130838.1(UBE3A):c.1373C>T (p.Pro458Leu)	UBE3A	15	25370741	Angelman syndrome
NM_130838.1(UBE3A):c.961C>T (p.Gln321Ter)	UBE3A	15	25371153	Angelman syndrome
NM_130838.1(UBE3A):c.1201C>T (p.Arg401Ter)	UBE3A	15	25370913	Angelman syndrome
NM_130838.1(UBE3A):c.1730G>A (p.Trp577Ter)	UBE3A	15	25356860	Angelman syndrome
NM_130838.1(UBE3A):c.1972C>T (p.Gln658Ter)	UBE3A	15	25355984	Angelman syndrome
NM_130838.1(UBE3A):c.2233C>T (p.Gln745Ter)	UBE3A	15	25354414	Angelman syndrome
NM_130838.1(UBE3A):c.1633G>A (p.Gly545Arg)	UBE3A	15	25360443	Angelman syndrome
NM_130838.1(UBE3A):c.1967C>T (p.Thr656Ile)	UBE3A	15	25355989	Angelman syndrome
NM_130838.1(UBE3A):c.2480C>T (p.Pro827Leu)	UBE3A	15	25339216	Angelman syndrome
NM_130838.1(UBE3A):c.964C>T (p.Gln322Ter)	UBE3A	15	25371150	Angelman syndrome
NM_001110792.1(MECP2):c.916C>T (p.Arg306Ter)	MECP2	X	154030948	Angelman syndrome;Autism, susceptibility to, X-linked 3;Mental retardation, X-linked, syndromic 13;Rett syndrome;not provided
NM_004992.3(MECP2):c.473C>T (p.Thr158Met)	MECP2	X	154031355	Angelman syndrome;Autism, susceptibility to, X-linked 3;Mental retardation, X-linked, syndromic 13;Rett syndrome;Severe neonatal-onset encephalopathy with microcephaly;not provided
NM_004992.3(MECP2):c.808C>T (p.Arg270Ter)	MECP2	X	154031020	Angelman syndrome;Encephalopathy, neonatal severeMental retardation, X-linked, syndromic 13Rett syndrome;Mental retardation, X-linked, syndromic 13;Rett syndrome;Severe neonatal-onset encephalopathy with microcephaly;not provided
NM_004992.3(MECP2):c.916C>T (p.Arg306Cys)	MECP2	X	154030912	Angelman syndrome;Intellectual disability;Mental retardation, X-linked, syndromic 13;Rett syndrome;not provided
NM_001845.5(COL4A1):c.1555G>A (p.Gly519Arg)	COL4A1	13	110187311	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps
NM_001845.5(COL4A1):c.1583G>A (p.Gly528Glu)	COL4A1	13	110187283	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps

NM_001845.5(COL4A1):c.1528G>A (p.Gly510Arg)	COL4A1	13	110192222	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps;Retinal arteries, tortuosity of
NM_000789.3(ACE):c.3683C>T (p.Pro1228Leu)	ACE	17	63496977	Angiotensin i-converting enzyme, benign serum increase
NM_002223.3(ITPR2):c.7492G>A (p.Gly2498Ser)	ITPR2	12	26400166	Anhidrosis, isolated, with normal sweat glands
NM_000280.4(PAX6):c.406C>T (p.Gln136Ter)	PAX6	11	31800808	Aniridia 1
NM_000280.4(PAX6):c.771G>A (p.Trp257Ter)	PAX6	11	31793797	Aniridia 1
NM_000280.4(PAX6):c.112C>T (p.Arg38Trp)	PAX6	11	31802733	Aniridia 1
NM_000280.4(PAX6):c.307C>T (p.Arg103Ter)	PAX6	11	31801611	Aniridia 1;not provided
NM_000280.4(PAX6):c.607C>T (p.Arg203Ter)	PAX6	11	31794705	Aniridia 1;not provided
NM_000280.4(PAX6):c.718C>T (p.Arg240Ter)	PAX6	11	31794079	Aniridia 1;not provided
NM_000280.4(PAX6):c.357+1G>A	PAX6	11	31801560	Aniridia 1;not provided
NM_017583.5(TRIM44):c.463G>A (p.Gly155Arg)	TRIM44	11	35663574	ANIRIDIA 3
NM_001029871.3(RSPO4):c.353G>A (p.Cys118Tyr)	RSPO4	20	967230	Anonychia
NM_001029871.3(RSPO4):c.218G>A (p.Cys73Tyr)	RSPO4	20	968000	Anonychia
NM_001029871.3(RSPO4):c.301C>T (p.Gln101Ter)	RSPO4	20	967282	Anonychia
NM_001029871.3(RSPO4):c.190C>T (p.Arg64Cys)	RSPO4	20	968028	Anonychia
NM_182894.2(VSX2):c.667G>A (p.Gly223Arg)	VSX2	14	74259689	Anophthalmia - microphthalmia
NM_022137.5(SMOC1):c.718C>T (p.Gln240Ter)	SMOC1	14	70010807	Anophthalmos with limb anomalies
NM_022137.5(SMOC1):c.664+1G>A	SMOC1	14	69994481	Anophthalmos with limb anomalies
NM_000503.5(EYA1):c.1087G>A (p.Glu363Lys)	EYA1	8	71244656	Anterior segment anomalies
NM_000104.3(CYP1B1):c.171G>A (p.Trp57Ter)	CYP1B1	2	38075218	ANTERIOR SEGMENT DYSGENESIS 6;CYP1B1-Related Disorders;Glaucoma, congenital;Irido-corneo-trabecular dysgenesis
NM_015692.2(CPAMD8):c.4549-1G>A	CPAMD8	19	16903624	ANTERIOR SEGMENT DYSGENESIS 8
NM_001243226.2(TCF4):c.806-1G>A	TCF4	18	55350409	Anteverted nares;Drooling;Epicanthus;Feeding difficulties in infancy;Global developmental delay;Short nose;Stereotypy
NM_000934.3(SERPINF2):c.1231G>A (p.Val411Met)	SERPINF2	17	1754289	Anti-plasmin deficiency, congenital
NM_000488.3(SERPINC1):c.1306G>A (p.Ala436Thr)	SERPINC1	1	173903978	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.235C>T (p.Arg79Cys)	SERPINC1	1	173914726	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.1277C>T (p.Ser426Leu)	SERPINC1	1	173904007	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.1273C>T (p.Arg425Cys)	SERPINC1	1	173904011	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.1316C>T (p.Pro439Leu)	SERPINC1	1	173903968	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.1274G>A (p.Arg425His)	SERPINC1	1	173904010	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.1240G>A (p.Ala414Thr)	SERPINC1	1	173904044	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.481C>T (p.Arg161Ter)	SERPINC1	1	173911942	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.1382C>T (p.Pro461Leu)	SERPINC1	1	173903902	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.1271G>A (p.Gly424Asp)	SERPINC1	1	173904013	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.391C>T (p.Leu131Phe)	SERPINC1	1	173914570	Antithrombin III deficiency
NM_000488.3(SERPINC1):c.166C>T (p.Arg56Cys)	SERPINC1	1	173914795	Antithrombin III deficiency;not provided
NM_000941.2(POR):c.731+1G>A	POR	7	75981607	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
NM_000941.2(POR):c.1370G>A (p.Arg457His)	POR	7	75985179	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis;Disordered steroidogenesis due to cytochrome p450 oxidoreductase deficiency
NM_000941.2(POR):c.1615G>A (p.Gly539Arg)	POR	7	75985795	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis;Disordered steroidogenesis due to cytochrome p450 oxidoreductase deficiency
NM_000138.4(FBN1):c.4588C>T (p.Arg1530Cys)	FBN1	15	48468097	Aortic aneurysm, ascending, and dissection;Ectopia lentis, isolated, autosomal dominant;Marfan syndrome;Thoracic aortic aneurysm and aortic dissection

NM_002317.6(LOX):c.125G>A (p.Trp42Ter)	LOX	5	122077861	AORTIC ANEURYSM, FAMILIAL THORACIC 10
NM_012186.2(FOXE3):c.410G>A (p.Gly137Asp)	FOXE3	1	47416725	AORTIC ANEURYSM, FAMILIAL THORACIC 11, SUSCEPTIBILITY TO
NM_022844.2(MYH11):c.2135G>A (p.Arg712Gln)	MYH11	16	15748092	Aortic aneurysm, familial thoracic 4;Thoracic aortic aneurysm and aortic dissection
NM_001613.2(ACTA2):c.536G>A (p.Arg179His)	ACTA2	10	88941309	Aortic aneurysm, familial thoracic 6;Connective tissue disorder;Moyamoya disease 5;Multisystemic smooth muscle dysfunction syndrome;Thoracic aortic aneurysm and aortic dissection;alterations of great arteries and veins;not provided
NM_001613.2(ACTA2):c.773G>A (p.Arg258His)	ACTA2	10	88939542	Aortic aneurysm, familial thoracic 6;Moyamoya disease 5;not provided
NM_001613.2(ACTA2):c.772C>T (p.Arg258Cys)	ACTA2	10	88939543	Aortic aneurysm, familial thoracic 6;Moyamoya disease 5;Thoracic aortic aneurysm and aortic dissection
NM_001613.2(ACTA2):c.353G>A (p.Arg118Gln)	ACTA2	10	88943813	Aortic aneurysm, familial thoracic 6;not provided
NM_001613.2(ACTA2):c.445C>T (p.Arg149Cys)	ACTA2	10	88941794	Aortic aneurysm, familial thoracic 6;Thoracic aortic aneurysm and aortic dissection
NM_001141945.2(ACTA2):c.115C>T (p.Arg39Cys)	ACTA2	10	88948816	Aortic aneurysm, familial thoracic 6;Thoracic aortic aneurysm and aortic dissection
NM_053025.3(MYLK):c.4438C>T (p.Arg1480Ter)	MYLK	3	123647405	Aortic aneurysm, familial thoracic 7
NM_001098512.2(PRKG1):c.530G>A (p.Arg177Gln)	PRKG1	10	51467819	Aortic aneurysm, familial thoracic 8;Thoracic aortic aneurysm and aortic dissection
NM_003480.3(MFAP5):c.472C>T (p.Arg158Ter)	MFAP5	12	8648141	Aortic aneurysm, familial thoracic 9
NM_000138.4(FBN1):c.5788+5G>A	FBN1	15	48446701	Aortic root dilatation;Arachnodactyly;Dissecting aortic aneurysm;Ectopia lentis;Ischemic stroke;Marfan syndrome;Melanoma;Severe Myopia;not provided
NM_005585.4(SMAD6):c.1244C>T (p.Pro415Leu)	SMAD6	15	66781288	Aortic valve disease 2
NM_000192.3(TBX5):c.443G>A (p.Trp148Ter)	TBX5	12	114398640	Aortic valve disease 2
NM_000192.3(TBX5):c.709C>T (p.Arg237Trp)	TBX5	12	114385522	Aortic valve disease 2;Heart, malformation of;Holt-Oram syndrome
NM_000192.3(TBX5):c.710G>A (p.Arg237Gln)	TBX5	12	114385521	Aortic valve disease 2;Holt-Oram syndrome;not provided
NM_000192.3(TBX5):c.835C>T (p.Arg279Ter)	TBX5	12	114366312	Aortic valve disease 2;not provided
NM_017617.4(NOTCH1):c.3319C>T (p.Arg1107Ter)	NOTCH1	9	136508238	Aortic valve disorder
NM_014753.3(BMS1):c.2789G>A (p.Arg930His)	BMS1	10	42820527	Aplasia cutis congenita
NM_198253.2(TERT):c.2147C>T (p.Ala716Val)	TERT	5	1278780	Aplastic anemia
NR_001566.1(TERC):n.178G>A	TERC	3	169764883	Aplastic anemia
NR_001566.1(TERC):n.180C>T	TERC	3	169764881	Aplastic anemia
NR_001566.1(TERC):n.305G>A	TERC	3	169764756	Aplastic anemia
NR_001566.1(TERC):n.322G>A	TERC	3	169764739	Aplastic anemia
NR_001566.1(TERC):n.323C>T	TERC	3	169764738	Aplastic anemia
NM_198253.2(TERT):c.2935C>T (p.Arg979Trp)	TERT	5	1260509	Aplastic anemia;Dyskeratosis congenita autosomal dominant
NM_198253.2(TERT):c.2080G>A (p.Val694Met)	TERT	5	1279341	Aplastic anemia;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1
NM_198253.2(TERT):c.3268G>A (p.Val1090Met)	TERT	5	1254395	Aplastic anemia;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1
NM_000039.2(APOA1):c.322C>T (p.Gln108Ter)	APOA1	11	116836290	Apolipoprotein A-I deficiency
NM_000196.3(HSD11B2):c.622C>T (p.Arg208Cys)	HSD11B2	16	67436100	Apparent mineralocorticoid excess
NM_000196.3(HSD11B2):c.637C>T (p.Arg213Cys)	HSD11B2	16	67436115	Apparent mineralocorticoid excess
NM_000196.3(HSD11B2):c.1009C>T (p.Arg337Cys)	HSD11B2	16	67436794	Apparent mineralocorticoid excess
NM_000196.3(HSD11B2):c.623G>A (p.Arg208His)	HSD11B2	16	67436101	Apparent mineralocorticoid excess
NM_000196.3(HSD11B2):c.835C>T (p.Arg279Cys)	HSD11B2	16	67436620	Apparent mineralocorticoid excess
NM_000196.3(HSD11B2):c.667G>A (p.Asp223Asn)	HSD11B2	16	67436251	Apparent mineralocorticoid excess
NM_000196.3(HSD11B2):c.664+14C>T	HSD11B2	16	67436156	Apparent mineralocorticoid excess
NM_000196.3(HSD11B2):c.680C>T (p.Pro227Leu)	HSD11B2	16	67436264	Apparent mineralocorticoid excess, mild
NM_198098.3(AQP1):c.113C>T (p.Pro38Leu)	AQP1	7	30912022	Aquaporin 1 deficiency
NM_000045.3(ARG1):c.871C>T (p.Arg291Ter)	ARG1	6	131583810	Arginase deficiency
NM_000045.3(ARG1):c.57+1G>A	ARG1	6	131573340	Arginase deficiency

NM_000045.3(ARG1):c.365G>A (p.Trp122Ter)	ARG1	6	131581278	Arginase deficiency;not provided
NM_000045.3(ARG1):c.61C>T (p.Arg21Ter)	ARG1	6	131576666	Arginase deficiency;not provided
NM_001482.2(GATM):c.446G>A (p.Trp149Ter)	GATM	15	45369364	Arginine:glycine amidinotransferase deficiency
NM_001482.2(GATM):c.505C>T (p.Arg169Ter)	GATM	15	45368240	Arginine:glycine amidinotransferase deficiency
NM_000048.3(ASL):c.283C>T (p.Arg95Cys)	ASL	7	66082443	Argininosuccinate lyase deficiency
NM_001024943.1(ASL):c.1153C>T (p.Arg385Cys)	ASL	7	66092566	Argininosuccinate lyase deficiency
NM_000048.3(ASL):c.532G>A (p.Val178Met)	ASL	7	66086751	Argininosuccinate lyase deficiency
NM_001024943.1(ASL):c.346C>T (p.Gln116Ter)	ASL	7	66082934	Argininosuccinate lyase deficiency
NM_000048.3(ASL):c.545G>A (p.Arg182Gln)	ASL	7	66086764	Argininosuccinate lyase deficiency
NM_000048.3(ASL):c.175G>A (p.Glu59Lys)	ASL	7	66081965	Argininosuccinate lyase deficiency
NM_000048.3(ASL):c.718+5G>A	ASL	7	66087796	Argininosuccinate lyase deficiency
NM_000048.3(ASL):c.889C>T (p.Arg297Trp)	ASL	7	66089146	Argininosuccinate lyase deficiency
NM_000048.3(ASL):c.1360C>T (p.Gln454Ter)	ASL	7	66092877	Argininosuccinate lyase deficiency
NM_000048.3(ASL):c.1135C>T (p.Arg379Cys)	ASL	7	66092078	Argininosuccinate lyase deficiency;not provided
NM_000048.3(ASL):c.1060C>T (p.Gln354Ter)	ASL	7	66089693	Argininosuccinate lyase deficiency;not provided
NM_000048.3(ASL):c.35G>A (p.Arg12Gln)	ASL	7	66081825	Argininosuccinate lyase deficiency;not provided
NM_000048.3(ASL):c.446+1G>A	ASL	7	66083175	Argininosuccinate lyase deficiency;not provided
NM_000048.3(ASL):c.544C>T (p.Arg182Ter)	ASL	7	66086763	Argininosuccinate lyase deficiency;not provided
NM_015295.2(SMCHD1):c.404G>A (p.Ser135Asn)	SMCHD1	18	2667011	Arhinia choanal atresia microphthalmia
NM_015295.2(SMCHD1):c.410G>A (p.Gly137Glu)	SMCHD1	18	2667017	Arhinia choanal atresia microphthalmia;Facioscapulohumeral muscular dystrophy 2
NM_031226.2(CYP19A1):c.1303C>T (p.Arg435Cys)	CYP19A1	15	51211017	Aromatase deficiency
NM_031226.2(CYP19A1):c.1310G>A (p.Cys437Tyr)	CYP19A1	15	51211010	Aromatase deficiency
NM_031226.2(CYP19A1):c.1123C>T (p.Arg375Cys)	CYP19A1	15	51212460	Aromatase deficiency
NM_031226.2(CYP19A1):c.296+1G>A	CYP19A1	15	51236858	Aromatase deficiency
NM_031226.2(CYP19A1):c.1094G>A (p.Arg365Gln)	CYP19A1	15	51212489	Aromatase deficiency
NM_031226.2(CYP19A1):c.628G>A (p.Glu210Lys)	CYP19A1	15	51222349	Aromatase deficiency
NM_001943.4(DSG2):c.1520G>A (p.Cys507Tyr)	DSG2	18	31536298	Arrhythmogenic right ventricular cardiomyopathy, type 10
NM_001943.4(DSG2):c.1750C>T (p.Gln584Ter)	DSG2	18	31538849	Arrhythmogenic right ventricular cardiomyopathy, type 10
NM_001943.4(DSG2):c.918G>A (p.Trp306Ter)	DSG2	18	31524792	Arrhythmogenic right ventricular cardiomyopathy, type 10;not provided
NM_004415.3(DSP):c.8501G>A (p.Arg2834His)	DSP	6	7585763	Arrhythmogenic right ventricular cardiomyopathy, type 8
NM_004415.3(DSP):c.2602C>T (p.Gln868Ter)	DSP	6	7575460	Arrhythmogenic right ventricular cardiomyopathy, type 8
NM_004415.3(DSP):c.3337C>T (p.Arg1113Ter)	DSP	6	7579527	Arrhythmogenic right ventricular cardiomyopathy, type 8
NM_004415.3(DSP):c.4822C>T (p.Gln1608Ter)	DSP	6	7581012	Arrhythmogenic right ventricular cardiomyopathy, type 8
NM_004415.3(DSP):c.268C>T (p.Gln90Ter)	DSP	6	7555815	Arrhythmogenic right ventricular cardiomyopathy, type 8;not provided
NM_004572.3(PKP2):c.1489C>T (p.Arg497Ter)	PKP2	12	32843203	Arrhythmogenic right ventricular cardiomyopathy, type 9
NM_004572.3(PKP2):c.1844C>T (p.Ser615Phe)	PKP2	12	32822594	Arrhythmogenic right ventricular cardiomyopathy, type 9
NM_004572.3(PKP2):c.368G>A (p.Trp123Ter)	PKP2	12	32878512	Arrhythmogenic right ventricular cardiomyopathy, type 9;not provided
NM_001943.4(DSG2):c.146G>A (p.Arg49His)	DSG2	18	31519867	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 10;not provided
NM_001943.4(DSG2):c.137G>A (p.Arg46Gln)	DSG2	18	31519858	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 10;not provided
NM_024334.2(TMEM43):c.1073C>T (p.Ser358Leu)	TMEM43	3	14141665	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 5;not provided
NM_004415.3(DSP):c.939+1G>A	DSP	6	7565521	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 8;Cardiomyopathy;not provided

NM_004415.3(DSP):c.478C>T (p.Arg160Ter)	DSP	6	7559281	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 8;not provided
NM_004572.3(PKP2):c.2203C>T (p.Arg735Ter)	PKP2	12	32802499	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 9
NM_004572.3(PKP2):c.1170+1G>A	PKP2	12	32868926	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 9
NM_004572.3(PKP2):c.235C>T (p.Arg79Ter)	PKP2	12	32879021	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 9;Cardiovascular phenotype;not provided
NM_004572.3(PKP2):c.2489+1G>A	PKP2	12	32796108	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 9;not provided
NM_004572.3(PKP2):c.1613G>A (p.Trp538Ter)	PKP2	12	32841103	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 9;not provided
NM_004572.3(PKP2):c.1237C>T (p.Arg413Ter)	PKP2	12	32850907	Arrhythmogenic right ventricular cardiomyopathy;Arrhythmogenic right ventricular cardiomyopathy, type 9;not provided
NM_004415.3(DSP):c.2821C>T (p.Arg941Ter)	DSP	6	7576986	Arrhythmogenic right ventricular cardiomyopathy;Cardiac arrest;not provided
NM_004281.3(BAG3):c.652C>T (p.Arg218Trp)	BAG3	10	119672399	Arrhythmogenic right ventricular cardiomyopathy;Dilated cardiomyopathy 1HH
NM_004415.3(DSP):c.4531C>T (p.Gln1511Ter)	DSP	6	7580721	Arrhythmogenic right ventricular cardiomyopathy;not provided
NM_004572.3(PKP2):c.1132C>T (p.Gln378Ter)	PKP2	12	32868965	Arrhythmogenic right ventricular cardiomyopathy;not provided
NM_004572.3(PKP2):c.1688+1G>A	PKP2	12	32841027	Arrhythmogenic right ventricular cardiomyopathy;not provided
NM_004572.3(PKP2):c.1912C>T (p.Gln638Ter)	PKP2	12	32822526	Arrhythmogenic right ventricular cardiomyopathy;not provided
NM_004572.3(PKP2):c.1162C>T (p.Arg388Trp)	PKP2	12	32868935	Arrhythmogenic right ventricular cardiomyopathy;not provided
NM_004415.3(DSP):c.5851C>T (p.Arg1951Ter)	DSP	6	7583113	Arrhythmogenic right ventricular cardiomyopathy;not provided
NM_003239.4(TGFB3):c.-30G>A	TGFB3	14	75980923	Arrhythmogenic right ventricular dysplasia, familial 1
NM_003239.4(TGFB3):c.*495C>T	TGFB3	14	75958692	Arrhythmogenic right ventricular dysplasia, familial 1
NM_198056.2(SCN5A):c.6048G>A (p.Val2016=)	SCN5A	3	38550324	Arrhythmogenic right ventricular dysplasia/cardiomyopathy
NM_198056.2(SCN5A):c.4867C>T (p.Arg1623Ter)	SCN5A	3	38551505	Arrhythmogenic right ventricular dysplasia/cardiomyopathy;Long QT syndrome 3;Sick sinus syndrome 1, autosomal recessive;not provided
NM_001198903.1(YY1AP1):c.724C>T (p.Gln242Ter)	YY1AP1	1	155676562	Arterial occlusive disease, progressive, with hypertension, heart defects, bone fragility, and brachysyndactyly
NM_001198903.1(YY1AP1):c.664C>T (p.Gln222Ter)	YY1AP1	1	155676622	Arterial occlusive disease, progressive, with hypertension, heart defects, bone fragility, and brachysyndactyly
NM_030777.3(SLC2A10):c.510G>A (p.Trp170Ter)	SLC2A10	20	46725546	Arterial tortuosity syndrome
NM_030777.3(SLC2A10):c.394C>T (p.Arg132Trp)	SLC2A10	20	46725430	Arterial tortuosity syndrome
NM_030777.3(SLC2A10):c.737G>A (p.Gly246Glu)	SLC2A10	20	46725773	Arterial tortuosity syndrome
NM_030777.3(SLC2A10):c.1309G>A (p.Glu437Lys)	SLC2A10	20	46726884	Arterial tortuosity syndrome
NM_030777.3(SLC2A10):c.1330C>T (p.Arg444Ter)	SLC2A10	20	46726905	Arterial tortuosity syndrome
NM_030777.3(SLC2A10):c.1334G>A (p.Gly445Glu)	SLC2A10	20	46726909	Arterial tortuosity syndrome
NM_030777.3(SLC2A10):c.1411+1G>A (p.Val430_I470del)	SLC2A10	20	46726987	Arterial tortuosity syndrome
NM_030777.3(SLC2A10):c.691C>T (p.Arg231Trp)	SLC2A10	20	46725727	Arterial tortuosity syndrome;not provided
NM_030777.3(SLC2A10):c.685C>T (p.Arg229Ter)	SLC2A10	20	46725721	Arterial tortuosity syndrome;Thoracic aortic aneurysm and aortic dissection
NM_030777.3(SLC2A10):c.692G>A (p.Arg231Gln)	SLC2A10	20	46725728	Arterial tortuosity syndrome;Thoracic aortic aneurysm and aortic dissection
NM_003334.3(UBA1):c.1731C>T (p.Asn577=)	UBA1	X	47206103	Arthrogryposis multiplex congenita, distal, X-linked
NM_198569.2(ADGRG6):c.19C>T (p.Arg7Ter)	ADGRG6	6	142309560	Arthrogryposis multiplex congenita;Lethal congenital contracture syndrome 9
NM_018668.4(VPS33B):c.1594C>T (p.Arg532Ter)	VPS33B	15	90999963	Arthrogryposis renal dysfunction cholestasis syndrome

NM_018668.4(VPS33B):c.1312C>T (p.Arg438Ter)	VPS33B	15	91002143	Arthrogryposis renal dysfunction cholestasis syndrome
NM_018668.4(VPS33B):c.700+1G>A	VPS33B	15	91006949	Arthrogryposis renal dysfunction cholestasis syndrome
NM_004826.3(ECEL1):c.1252C>T (p.Arg418Cys)	ECEL1	2	232484156	Arthrogryposis, distal, type 5d
NM_004826.3(ECEL1):c.590G>A (p.Gly197Asp)	ECEL1	2	232486064	Arthrogryposis, distal, type 5d
NM_004826.3(ECEL1):c.2023G>A (p.Ala675Thr)	ECEL1	2	232481123	Arthrogryposis, distal, type 5d
NM_004826.3(ECEL1):c.1184G>A (p.Arg395Gln)	ECEL1	2	232484472	Arthrogryposis, distal, type 5d
NM_004826.3(ECEL1):c.1470G>A (p.Trp490Ter)	ECEL1	2	232483452	Arthrogryposis, distal, type 5d
NM_004826.3(ECEL1):c.997C>T (p.Arg333Ter)	ECEL1	2	232484863	Arthrogryposis, distal, type 5d;not provided
NM_022068.3(PIEZO2):c.5053C>T (p.Arg1685Ter)	PIEZO2	18	10715679	ARTHROGRYPOSIS, DISTAL, WITH IMPAIRED PROPRIOCEPTION AND TOUCH
NM_022068.3(PIEZO2):c.4723C>T (p.Arg1575Ter)	PIEZO2	18	10736621	ARTHROGRYPOSIS, DISTAL, WITH IMPAIRED PROPRIOCEPTION AND TOUCH
NM_001271684.1(SLC35A3):c.514C>T (p.Gln172Ter)	SLC35A3	1	100011413	Arthrogryposis, mental retardation, and seizures
NM_033116.5(NEK9):c.2042G>A (p.Arg681His)	NEK9	14	75097231	Arthrogryposis, perthes disease, and upward gaze palsy
NM_022067.3(VIPAS39):c.535C>T (p.Gln179Ter)	VIPAS39	14	77444311	Arthrogryposis, renal dysfunction, and cholestasis 2
NM_022067.3(VIPAS39):c.658C>T (p.Arg220Ter)	VIPAS39	14	77442636	Arthrogryposis, renal dysfunction, and cholestasis 2
NM_022067.3(VIPAS39):c.871C>T (p.Gln291Ter)	VIPAS39	14	77435885	Arthrogryposis, renal dysfunction, and cholestasis 2
NM_006757.3(TNNT3):c.188G>A (p.Arg63His)	TNNT3	11	1933737	Arthyrgryposis, distal, type 2B;Distal arthrogryposis type 2B;not provided
NM_006757.3(TNNT3):c.187C>T (p.Arg63Cys)	TNNT3	11	1933736	Arthyrgryposis, distal, type 2B;not provided
NM_022786.2(ARV1):c.294+1G>A	ARV1	1	230988440	ARV1-related condition;Epileptic encephalopathy, early infantile, 38
NM_000487.5(ARSA):c.511G>A (p.Asp171Asn)	ARSA	22	50627007	Arylsulfatase A pseudodeficiency
NM_000487.5(ARSA):c.1462C>T (p.Gln488Ter)	ARSA	22	50625213	Arylsulfatase A pseudodeficiency
NM_000487.5(ARSA):c.1150G>A (p.Glu384Lys)	ARSA	22	50625639	Arylsulfatase a pseudodeficiency, intermediate;Metachromatic leukodystrophy
NM_000487.5(ARSA):c.1136C>T (p.Pro379Leu)	ARSA	22	50625653	Arylsulfatase a pseudodeficiency, severe
NM_000027.3(AGA):c.904G>A (p.Gly302Arg)	AGA	4	177433250	Aspartylglycosaminuria
NM_000027.3(AGA):c.179G>A (p.Gly60Asp)	AGA	4	177440375	Aspartylglycosaminuria
NM_000027.3(AGA):c.302C>T (p.Ala101Val)	AGA	4	177439668	Aspartylglycosaminuria
NM_000027.3(AGA):c.319C>T (p.Arg107Ter)	AGA	4	177439651	Aspartylglycosaminuria
NM_000027.3(AGA):c.677G>A (p.Gly226Asp)	AGA	4	177436297	Aspartylglycosaminuria;not provided
NM_018977.3(NLGN3):c.1351C>T (p.Arg451Cys)	NLGN3	X	71167508	Asperger syndrome X-linked 1;Autism, susceptibility to, X-linked 1
NM_020800.2(IFT80):c.721G>A (p.Gly241Arg)	IFT80	3	160356069	Asphyxiating thoracic dystrophy 2
NM_024753.4(TTC21B):c.1231C>T (p.Arg411Ter)	TTC21B	2	165929290	Asphyxiating thoracic dystrophy 4
NM_002295.5(RPSA):c.25C>T (p.Gln9Ter)	RPSA	3	39407678	Asplenia, isolated congenital
NM_002295.5(RPSA):c.538C>T (p.Arg180Trp)	RPSA	3	39411688	Asplenia, isolated congenital
NM_002295.5(RPSA):c.556C>T (p.Arg186Cys)	RPSA	3	39411706	Asplenia, isolated congenital
NM_002295.5(RPSA):c.172C>T (p.Leu58Phe)	RPSA	3	39408644	Asplenia, isolated congenital
NM_006895.2(HNMT):c.314C>T (p.Thr105Ile)	HNMT	2	138002079	Asthma, susceptibility to
NM_007199.2(IRAK3):c.227G>A (p.Trp76Ter)	IRAK3	12	66203804	Asthma-related traits, susceptibility to, 5
NM_000834.3(GRIN2B):c.2002G>A (p.Asp668Asn)	GRIN2B	12	13608611	Astigmatism;Delayed speech and language development;Joint hypermobility;Long fingers;Motor delay
NM_002834.4(PTPN11):c.226G>A (p.Glu76Lys)	PTPN11	12	112450406	Astrocytoma;Colorectal Neoplasms;Juvenile myelomonocytic leukemia;Multiple myeloma;Neuroblastoma;Noonan syndrome;Rasopathy;Squamous cell carcinoma of lung;not provided
NM_000370.3(TTPA):c.175C>T (p.Arg59Trp)	TTPA	8	63085847	Ataxia with vitamin E deficiency
NM_000370.3(TTPA):c.358G>A (p.Ala120Thr)	TTPA	8	63072935	Ataxia with vitamin E deficiency
NM_000370.3(TTPA):c.421G>A (p.Glu141Lys)	TTPA	8	63066035	Ataxia with vitamin E deficiency
NM_000370.3(TTPA):c.661C>T (p.Arg221Trp)	TTPA	8	63064208	Ataxia with vitamin E deficiency
NM_000370.3(TTPA):c.575G>A (p.Arg192His)	TTPA	8	63064294	Ataxia with vitamin E deficiency;Ataxia, Friedreich-like, with isolated vitamin E deficiency
NM_000370.3(TTPA):c.400C>T (p.Arg134Ter)	TTPA	8	63066056	Ataxia with vitamin E deficiency;Ataxia, Friedreich-like, with isolated vitamin E deficiency

m.1606G>A	MT-TV	MT	1606	Ataxia, progressive seizures, mental deterioration, and hearing loss
NM_001160223.1(RNF170):c.595C>T (p.Arg199Cys)	RNF170	8	42856341	Ataxia, sensory, autosomal dominant
NM_006612.5(KIF1C):c.2191C>T (p.Arg731Ter)	KIF1C	17	5022272	Ataxia, spastic, 2, autosomal recessive
NM_006612.5(KIF1C):c.505C>T (p.Arg169Trp)	KIF1C	17	5002539	Ataxia, spastic, 2, autosomal recessive
NM_006612.5(KIF1C):c.183G>A (p.Ser61=)	KIF1C	17	5000848	Ataxia, spastic, 2, autosomal recessive
NM_000271.4(NPC1):c.1421C>T (p.Pro474Leu)	NPC1	18	23554890	Ataxia;Cataplexy;Cognitive impairment;Headache;Postural instability;Speech apraxia
NM_001005463.2(EBF3):c.530C>T (p.Pro177Leu)	EBF3	10	129957282	Ataxia;Delayed speech and language development;HYPOTONIA, ATAXIA, AND DELAYED DEVELOPMENT SYNDROME;Intellectual disability (moderate);Muscular hypotonia
NM_001005463.2(EBF3):c.488G>A (p.Arg163Gln)	EBF3	10	129957324	Ataxia;Expressive language delay;Global developmental delay;HYPOTONIA, ATAXIA, AND DELAYED DEVELOPMENT SYNDROME;Intellectual disability;Muscular hypotonia
NM_001142633.2(PIK3R5):c.1885C>T (p.Pro629Ser)	PIK3R5	17	8887116	Ataxia-oculomotor apraxia 3
NM_000051.3(ATM):c.5188C>T (p.Arg1730Ter)	ATM	11	108301658	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.8307G>A (p.Trp2769Ter)	ATM	11	108343260	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.7240C>T (p.Gln2414Ter)	ATM	11	108329171	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.1369C>T (p.Arg457Ter)	ATM	11	108250834	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.1240C>T (p.Gln414Ter)	ATM	11	108250705	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.5249G>A (p.Trp1750Ter)	ATM	11	108301719	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.6272G>A (p.Trp2091Ter)	ATM	11	108317446	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.2554C>T (p.Gln852Ter)	ATM	11	108267258	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.4906C>T (p.Gln1636Ter)	ATM	11	108295056	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.6658C>T (p.Gln2220Ter)	ATM	11	108325395	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.8880G>A (p.Trp2960Ter)	ATM	11	108365111	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.4110-1G>A	ATM	11	108288976	Ataxia-telangiectasia syndrome
NM_000051.3(ATM):c.9139C>T (p.Arg3047Ter)	ATM	11	108365476	Ataxia-telangiectasia syndrome;Ataxia-telangiectasia without immunodeficiency;not provided
NM_000051.3(ATM):c.8977C>T (p.Arg2993Ter)	ATM	11	108365208	Ataxia-telangiectasia syndrome;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.8185C>T (p.Gln2729Ter)	ATM	11	108335878	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.7951C>T (p.Gln2651Ter)	ATM	11	108333909	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.901+1G>A	ATM	11	108245027	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.8293G>A (p.Gly2765Ser)	ATM	11	108343246	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.802C>T (p.Gln268Ter)	ATM	11	108244927	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.2413C>T (p.Arg805Ter)	ATM	11	108259022	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.331+5G>A	ATM	11	108229328	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.5798G>A (p.Trp1933Ter)	ATM	11	108310195	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.6397C>T (p.Gln2133Ter)	ATM	11	108320003	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.4396C>T (p.Arg1466Ter)	ATM	11	108289761	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.103C>T (p.Arg35Ter)	ATM	11	108227806	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.3576G>A (p.Lys1192=)	ATM	11	108281168	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.7327C>T (p.Arg2443Ter)	ATM	11	108330233	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.1339C>T (p.Arg447Ter)	ATM	11	108250804	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.170G>A (p.Trp57Ter)	ATM	11	108227873	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.6100C>T (p.Arg2034Ter)	ATM	11	108316015	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.6572+1G>A	ATM	11	108321421	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.7456C>T (p.Arg2486Ter)	ATM	11	108330362	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.8494C>T (p.Arg2832Cys)	ATM	11	108345818	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided

NM_000051.3(ATM):c.8786+1G>A	ATM	11	108353881	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.7788G>A (p.Glu2596=)	ATM	11	108332037	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.2921+1G>A	ATM	11	108271147	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.7913G>A (p.Trp2638Ter)	ATM	11	108332886	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.5908C>T (p.Gln1970Ter)	ATM	11	108310305	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.3993+1G>A	ATM	11	108284474	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.9022C>T (p.Arg3008Cys)	ATM	11	108365359	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.742C>T (p.Arg248Ter)	ATM	11	108244867	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.3049C>T (p.Gln1017Ter)	ATM	11	108271378	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.6095G>A (p.Arg2032Lys)	ATM	11	108315911	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.6679C>T (p.Arg2227Cys)	ATM	11	108325416	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.3G>A (p.Met1Ile)	ATM	11	108227627	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.5644C>T (p.Arg1882Ter)	ATM	11	108304822	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.748C>T (p.Arg250Ter)	ATM	11	108244873	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.4735C>T (p.Gln1579Ter)	ATM	11	108293436	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.2023C>T (p.Gln675Ter)	ATM	11	108253938	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.3078-1G>A	ATM	11	108272531	Ataxia-telangiectasia syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.3663G>A (p.Trp1221Ter)	ATM	11	108282796	Ataxia-telangiectasia syndrome;not provided
NM_000051.3(ATM):c.8988-1G>A	ATM	11	108365324	Ataxia-telangiectasia syndrome;not provided
NM_000051.3(ATM):c.1463G>A (p.Trp488Ter)	ATM	11	108250928	Ataxia-telangiectasia syndrome;not provided
NM_000051.3(ATM):c.8545C>T (p.Arg2849Ter)	ATM	11	108345869	Ataxia-telangiectasia syndrome;not provided;not specified
NM_000051.3(ATM):c.496+5G>A	ATM	11	108235839	Ataxia-telangiectasia variant;Hereditary cancer-predisposing syndrome;not provided
NM_005591.3(MRE11):c.1897C>T (p.Arg633Ter)	MRE11	11	94437206	Ataxia-telangiectasia-like disorder 1
NM_005591.3(MRE11):c.140C>T (p.Ala47Val)	MRE11	11	94490846	Ataxia-telangiectasia-like disorder 1
NM_005591.3(MRE11):c.1714C>T (p.Arg572Ter)	MRE11	11	94447288	Ataxia-telangiectasia-like disorder 1;Hereditary cancer-predisposing syndrome
NM_005591.3(MRE11):c.659+1G>A	MRE11	11	94476288	Ataxia-telangiectasia-like disorder 1;Hereditary cancer-predisposing syndrome
NM_000515.4(GH1):c.59G>A (p.Trp20Ter)	GH1	17	63918458	Ateleiotic dwarfism
NM_001457.3(FLNB):c.518C>T (p.Ala173Val)	FLNB	3	58077271	Ateliosteogenesis type 1
NM_001457.3(FLNB):c.517G>A (p.Ala173Thr)	FLNB	3	58077270	Ateliosteogenesis type 1
NM_000112.3(SLC26A2):c.764G>A (p.Gly255Glu)	SLC26A2	5	149980357	Ateliosteogenesis type 2
NM_000112.3(SLC26A2):c.2144C>T (p.Ala715Val)	SLC26A2	5	149981737	Ateliosteogenesis type 2
NM_000112.3(SLC26A2):c.835C>T (p.Arg279Trp)	SLC26A2	5	149980428	Ateliosteogenesis type 2;Diastrophic dysplasia;Multiple epiphyseal dysplasia 4;SLC26A2-Related Disorders;not provided
NM_001457.3(FLNB):c.602C>T (p.Ala201Val)	FLNB	3	58078777	Ateliosteogenesis type 3
NM_005522.4(HOXA1):c.76C>T (p.Arg26Ter)	HOXA1	7	27095837	Athabaskan brainstem dysgenesis
NM_001063.3(TF):c.229G>A (p.Asp77Asn)	TF	3	133753607	Atransferrinemia
NM_001063.3(TF):c.1180G>A (p.Glu394Lys)	TF	3	133759306	Atransferrinemia
NM_198056.2(SCN5A):c.3823G>A (p.Asp1275Asn)	SCN5A	3	38566426	Atrial fibrillation, familial, 10;Atrial standstill 1, digenic;Brugada syndrome;Dilated cardiomyopathy 1E;not provided
NM_005266.6(GJA5):c.145C>T (p.Gln49Ter)	GJA5	1	147759094	Atrial fibrillation, familial, 11
NM_005266.6(GJA5):c.253G>A (p.Val85Ile)	GJA5	1	147758986	Atrial fibrillation, familial, 11
NM_005691.3(ABCC9):c.4640C>T (p.Thr1547Ile)	ABCC9	12	21805184	Atrial fibrillation, familial, 12
NM_001037.4(SCN1B):c.254G>A (p.Arg85His)	SCN1B	19	35033545	Atrial fibrillation, familial, 13;not provided
NM_004588.4(SCN2B):c.83G>A (p.Arg28Gln)	SCN2B	11	118168739	Atrial fibrillation, familial, 14
NM_153485.2(NUP155):c.1172G>A (p.Arg391His)	NUP155	5	37341164	Atrial fibrillation, familial, 15

NM_018400.3(SCN3B):c.17G>A (p.Arg6Lys)	SCN3B	11	123653785	Atrial fibrillation, familial, 16
NM_000218.2(KCNQ1):c.686G>A (p.Gly229Asp)	KCNQ1	11	2572015	Atrial fibrillation, familial, 3;Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided
NM_000218.2(KCNQ1):c.692G>A (p.Arg231His)	KCNQ1	11	2572021	Atrial fibrillation, familial, 3;Congenital long QT syndrome;Long QT syndrome;Long QT syndrome 1;not provided
NM_002234.3(KCNA5):c.1727C>T (p.Ala576Val)	KCNA5	12	5045874	Atrial fibrillation, familial, 7
NM_002234.3(KCNA5):c.1828G>A (p.Glu610Lys)	KCNA5	12	5045975	Atrial fibrillation, familial, 7
NM_005266.6(GJA5):c.262C>T (p.Pro88Ser)	GJA5	1	147758977	Atrial fibrillation, somatic
NM_198056.2(SCN5A):c.1963G>A (p.Glu655Lys)	SCN5A	3	38598978	Atrial fibrillation;Atrial fibrillation, familial, 10;not provided
NM_000218.2(KCNQ1):c.421G>A (p.Val141Met)	KCNQ1	11	2527962	Atrial fibrillation;Long QT syndrome;Short QT syndrome 2
NM_002052.4(GATA4):c.886G>A (p.Gly296Ser)	GATA4	8	11750213	Atrial septal defect 2
NM_002052.4(GATA4):c.155C>T (p.Ser52Phe)	GATA4	8	11708467	Atrial septal defect 2
NM_002052.4(GATA4):c.839C>T (p.Thr280Met)	GATA4	8	11750166	Atrial septal defect 2
NM_001077653.2(TBX20):c.583C>T (p.Gln195Ter)	TBX20	7	35245020	Atrial septal defect 4
NM_004387.3(NKX2-5):c.533C>T (p.Thr178Met)	NKX2-5	5	173233011	Atrial septal defect 7 with or without atrioventricular conduction defects
NM_004387.3(NKX2-5):c.508C>T (p.Gln170Ter)	NKX2-5	5	173233036	Atrial septal defect 7 with or without atrioventricular conduction defects
NM_004387.3(NKX2-5):c.592C>T (p.Gln198Ter)	NKX2-5	5	173232952	Atrial septal defect 7 with or without atrioventricular conduction defects
NM_004387.3(NKX2-5):c.568C>T (p.Arg190Cys)	NKX2-5	5	173232976	Atrial septal defect 7 with or without atrioventricular conduction defects
NM_005257.5(GATA6):c.551G>A (p.Ser184Asn)	GATA6	18	22171695	Atrial septal defect 9;Tetralogy of Fallot
NM_006172.3(NPPA):c.449G>A (p.Arg150Gln)	NPPA	1	11847114	Atrial standstill 2
NM_005144.4(HR):c.1837C>T (p.Arg613Ter)	HR	8	22123727	Atrichia with papular lesions
NM_005144.4(HR):c.3526C>T (p.Gln1176Ter)	HR	8	22115744	Atrichia with papular lesions
NM_198056.2(SCN5A):c.4783G>A (p.Asp1595Asn)	SCN5A	3	38554309	Atrioventricular block;Brugada syndrome;Progressive familial heart block type 1A;not provided
NM_001031717.3(CRELD1):c.932C>T (p.Thr311Ile)	CRELD1	3	9943399	Atrioventricular septal defect 2
NM_001077415.2(CRELD1):c.1240G>A (p.Glu414Lys)	CRELD1	3	9944556	Atrioventricular septal defect 2
NM_002052.4(GATA4):c.487C>T (p.Pro163Ser)	GATA4	8	11708799	Atrioventricular septal defect 4;Tetralogy of Fallot;Ventricular septal defect 1
NM_005257.5(GATA6):c.533C>T (p.Ala178Val)	GATA6	18	22171677	Atrioventricular septal defect 5
NM_000165.4(GJA1):c.1127G>A (p.Arg376Gln)	GJA1	6	121447974	Atrioventricular septal defect and common atrioventricular junction;Hypoplastic left heart syndrome
NM_000266.3(NDP):c.287G>A (p.Cys96Tyr)	NDP	X	43949914	Atrophia bulborum hereditaria
NM_000266.3(NDP):c.181C>T (p.Leu61Phe)	NDP	X	43950020	Atrophia bulborum hereditaria
NM_000266.3(NDP):c.313G>A (p.Ala105Thr)	NDP	X	43949888	Atrophia bulborum hereditaria
NM_000266.3(NDP):c.302C>T (p.Ser101Phe)	NDP	X	43949899	Atrophia bulborum hereditaria
NM_000266.3(NDP):c.325C>T (p.Arg109Ter)	NDP	X	43949876	Atrophia bulborum hereditaria
NM_000266.3(NDP):c.220C>T (p.Arg74Cys)	NDP	X	43949981	Atrophia bulborum hereditaria;Familial exudative vitreoretinopathy, X-linked
NM_000489.4(ATRX):c.7156C>T (p.Arg2386Ter)	ATRX	X	77520832	ATR-X syndrome
NM_000489.4(ATRX):c.5225G>A (p.Arg1742Lys)	ATRX	X	77620442	ATR-X syndrome
NM_000489.4(ATRX):c.4862C>T (p.Thr1621Met)	ATRX	X	77633660	ATR-X syndrome
NM_000489.4(ATRX):c.6122G>A (p.Ser2041Asn)	ATRX	X	77589929	ATR-X syndrome
NM_000489.4(ATRX):c.109C>T (p.Arg37Ter)	ATRX	X	77717155	ATR-X syndrome;Intellectual disability;Mental retardation-hypotonic facies syndrome, X-linked
NM_000489.4(ATRX):c.6392G>A (p.Arg2131Gln)	ATRX	X	77558781	ATR-X syndrome;Mental retardation-hypotonic facies syndrome X-linked, 1;Mental retardation-hypotonic facies syndrome, X-linked
NM_006578.3(GNB5):c.242C>T (p.Ser81Leu)	GNB5	15	52153947	Attention deficit hyperactivity disorder;Delayed speech and language development;Global developmental delay;LANGUAGE DELAY AND ATTENTION DEFICIT-HYPERACTIVITY DISORDER/COGNITIVE IMPAIRMENT WITH OR WITHOUT CARDIAC ARRHYTHMIA
NM_001039803.2(CDK20):c.564G>A (p.Trp188Ter)	CDK20	9	87969919	Attention deficit hyperactivity disorder;Hearing impairment;Obsessive-compulsive behavior;Seizures

NM_173353.3(TPH2):c.907C>T (p.Arg303Trp)	TPH2	12	71979053	Attention deficit-hyperactivity disorder 7
NM_000186.3(CFH):c.3572C>T (p.Ser1191Leu)	CFH	1	196747189	Atypical hemolytic-uremic syndrome 1
NM_002389.4(CD46):c.175C>T (p.Arg59Ter)	CD46	1	207757091	Atypical hemolytic-uremic syndrome 2
NM_002389.4(CD46):c.104G>A (p.Cys35Tyr)	CD46	1	207757020	Atypical hemolytic-uremic syndrome 2
NM_000204.4(CFI):c.1420C>T (p.Arg474Ter)	CFI	4	109746231	Atypical hemolytic-uremic syndrome 3
NM_000204.4(CFI):c.1637G>A (p.Trp546Ter)	CFI	4	109741008	Atypical hemolytic-uremic syndrome 3
NM_000204.4(CFI):c.949C>T (p.Arg317Trp)	CFI	4	109749594	Atypical hemolytic-uremic syndrome 3
NM_000204.4(CFI):c.1555G>A (p.Asp519Asn)	CFI	4	109741090	Atypical hemolytic-uremic syndrome 3
NM_000064.3(C3):c.1775G>A (p.Arg592Gln)	C3	19	6709754	Atypical hemolytic-uremic syndrome 5
NM_000064.3(C3):c.3281C>T (p.Ala1094Val)	C3	19	6693033	Atypical hemolytic-uremic syndrome 5
NM_000064.3(C3):c.3343G>A (p.Asp1115Asn)	C3	19	6692971	Atypical hemolytic-uremic syndrome 5
NM_000361.2(THBD):c.1483C>T (p.Pro495Ser)	THBD	20	23048022	Atypical hemolytic-uremic syndrome 6
NM_003159.2(CDKL5):c.352C>T (p.Gln118Ter)	CDKL5	X	18579917	Atypical Rett syndrome
NM_003159.2(CDKL5):c.403+1G>A	CDKL5	X	18579969	Atypical Rett syndrome
NM_003159.2(CDKL5):c.464-1G>A	CDKL5	X	18584262	Atypical Rett syndrome
NM_003159.2(CDKL5):c.1375C>T (p.Gln459Ter)	CDKL5	X	18604299	Atypical Rett syndrome
NM_003159.2(CDKL5):c.2046+1G>A	CDKL5	X	18608913	Atypical Rett syndrome
NM_003159.2(CDKL5):c.2704C>T (p.Gln902Ter)	CDKL5	X	18628578	Atypical Rett syndrome
NM_003159.2(CDKL5):c.2500C>T (p.Gln834Ter)	CDKL5	X	18628374	Atypical Rett syndrome;Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.119C>T (p.Ala40Val)	CDKL5	X	18564496	Atypical Rett syndrome;Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.175C>T (p.Arg59Ter)	CDKL5	X	18575383	Atypical Rett syndrome;Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.1648C>T (p.Arg550Ter)	CDKL5	X	18604572	Atypical Rett syndrome;Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.1675C>T (p.Arg559Ter)	CDKL5	X	18604599	Atypical Rett syndrome;Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.532C>T (p.Arg178Trp)	CDKL5	X	18584331	Atypical Rett syndrome;Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.2047-1G>A	CDKL5	X	18609464	Atypical Rett syndrome;not provided
NM_003159.2(CDKL5):c.463+1G>A	CDKL5	X	18581951	Atypical Rett syndrome;not provided
NM_002140.4(HNRNPK):c.257G>A (p.Arg86His)	HNRNPK	9	83975462	AU-KLINE SYNDROME;not provided
NM_005786.5(TSHZ1):c.723G>A (p.Trp241Ter)	TSHZ1	18	75286265	Aural atresia, congenital
NM_000933.3(PLCB4):c.1862G>A (p.Arg621His)	PLCB4	20	9409080	Auriculocondylar syndrome 1;Auriculocondylar syndrome 2
NM_000933.3(PLCB4):c.1861C>T (p.Arg621Cys)	PLCB4	20	9409079	Auriculocondylar syndrome 1;Auriculocondylar syndrome 2
NM_001172646.1(PLCB4):c.1078G>A (p.Asp360Asn)	PLCB4	20	9387476	Auriculocondylar syndrome 2
NM_173653.3(SLC9A9):c.1267C>T (p.Arg423Ter)	SLC9A9	3	143493701	Autism 16
NM_024757.4(EHMT1):c.3413G>A (p.Trp1138Ter)	EHMT1	9	137817477	Autism spectrum disorders;Chromosome 9q deletion syndrome
NM_020920.3(CHD8):c.2875C>T (p.Gln959Ter)	CHD8	14	21403019	Autism, susceptibility to, 18
NM_020920.3(CHD8):c.3172C>T (p.Arg1058Ter)	CHD8	14	21402010	Autism, susceptibility to, 18
NM_181332.2(NLGN4X):c.301C>T (p.Arg101Ter)	NLGN4X	X	6151166	Autism, susceptibility to, X-linked 2
NM_018094.4(GSPT2):c.1021G>A (p.Val341Ile)	GSPT2	X	51744647	Autism;Delayed speech and language development;Seizures
NM_001199922.1(SIAE):c.830C>T (p.Thr277Met)	SIAE	11	124647396	Autoimmune disease 6
NM_139276.2(STAT3):c.2107G>A (p.Ala703Thr)	STAT3	17	42317219	Autoimmune disease, multisystem, infantile-onset, 1
NM_139276.2(STAT3):c.1988C>T (p.Thr663Ile)	STAT3	17	42322395	Autoimmune disease, multisystem, infantile-onset, 1
NM_139276.2(STAT3):c.1261G>A (p.Gly421Arg)	STAT3	17	42329430	Autoimmune disease, multisystem, infantile-onset, 1
NM_139276.2(STAT3):c.454C>T (p.Arg152Trp)	STAT3	17	42339328	Autoimmune disease, multisystem, infantile-onset, 1
NM_139276.2(STAT3):c.1243G>A (p.Glu415Lys)	STAT3	17	42329448	Autoimmune disease, multisystem, infantile-onset, 1;Hyperimmunoglobulin E syndrome
NM_139276.2(STAT3):c.2147C>T (p.Thr716Met)	STAT3	17	42316899	Autoimmune disease, multisystem, infantile-onset, 1;not provided
NM_001079.3(ZAP70):c.574C>T (p.Arg192Trp)	ZAP70	2	97732893	Autoimmune disease, multisystem, infantile-onset, 2;Combined immunodeficiency

NM_001098398.1(COPA):c.698G>A (p.Arg233His)	COPA	1	160323439	Autoimmune interstitial lung, joint, and kidney disease
NM_001098398.1(COPA):c.721G>A (p.Glu241Lys)	COPA	1	160314111	Autoimmune interstitial lung, joint, and kidney disease
NM_000043.5(FAS):c.817C>T (p.Gln273Ter)	FAS	10	89014259	Autoimmune lymphoproliferative syndrome, type 1a
NM_000043.5(FAS):c.361C>T (p.Arg121Trp)	FAS	10	89008915	Autoimmune lymphoproliferative syndrome, type 1a
NM_000043.5(FAS):c.809C>T (p.Thr270Ile)	FAS	10	89014251	Autoimmune lymphoproliferative syndrome, type 1a
NM_000043.5(FAS):c.73G>A (p.Ala25Thr)	FAS	10	89003071	Autoimmune lymphoproliferative syndrome, type 1a
NM_032977.3(CASP10):c.853C>T (p.Leu285Phe)	CASP10	2	201208114	Autoimmune lymphoproliferative syndrome, type 2
NM_006254.3(PRKCD):c.1352+1G>A	PRKCD	3	53186696	Autoimmune lymphoproliferative syndrome, type III
NM_006254.3(PRKCD):c.1528G>A (p.Gly510Ser)	PRKCD	3	53188832	Autoimmune lymphoproliferative syndrome, type III
NM_006254.3(PRKCD):c.1840C>T (p.Arg614Trp)	PRKCD	3	53189969	Autoimmune lymphoproliferative syndrome, type III
NM_005214.4(CTLA4):c.151C>T (p.Arg51Ter)	CTLA4	2	203870627	Autoimmune lymphoproliferativE syndrome, type V
NM_005214.4(CTLA4):c.208C>T (p.Arg70Trp)	CTLA4	2	203870684	Autoimmune lymphoproliferativE syndrome, type V
NM_001033053.2(NLRP1):c.2176C>T (p.Arg726Trp)	NLRP1	17	5558520	AUTOINFLAMMATION WITH ARTHRITIS AND DYSKERATOSIS
NM_006290.3(TNFAIP3):c.811C>T (p.Arg271Ter)	TNFAIP3	6	137877081	Autoinflammatory syndrome, familial, Behcet-like
NM_001199107.1(TBC1D24):c.404C>T (p.Pro135Leu)	TBC1D24	16	2496552	Autosomal dominant epilepsy;Parkinsonism
NM_022336.3(EDAR):c.1259G>A (p.Arg420Gln)	EDAR	2	108896995	Autosomal dominant hypohidrotic ectodermal dysplasia;Ectodermal dysplasia 10a, hypohidrotic/hair/tooth type, autosomal dominant;not provided
NM_020638.2(FGF23):c.536G>A (p.Arg179Gln)	FGF23	12	4370563	Autosomal dominant hypophosphatemic rickets
NM_020638.2(FGF23):c.527G>A (p.Arg176Gln)	FGF23	12	4370572	Autosomal dominant hypophosphatemic rickets;not provided
NM_020638.2(FGF23):c.535C>T (p.Arg179Trp)	FGF23	12	4370564	Autosomal dominant hypophosphatemic rickets;not provided
NM_000515.4(GH1):c.291+1G>A	GH1	17	63918016	Autosomal dominant isolated somatotropin deficiency
NM_000515.4(GH1):c.291+2delG>A	GH1	17	63917989	Autosomal dominant isolated somatotropin deficiency
NM_000515.4(GH1):c.291+5G>A	GH1	17	63918012	Autosomal dominant isolated somatotropin deficiency
NM_000515.4(GH1):c.626G>A (p.Arg209His)	GH1	17	63917337	Autosomal dominant isolated somatotropin deficiency
NM_130837.2(OPA1):c.1817G>A (p.Cys606Tyr)	OPA1	3	193647127	Autosomal dominant optic atrophy plus syndrome
NM_015560.2(OPA1):c.1334G>A (p.Arg445His)	OPA1	3	193643996	Autosomal dominant optic atrophy plus syndrome;Dominant hereditary optic atrophy;not provided
NM_002693.2(POLG):c.1532G>A (p.Ser511Asn)	POLG	15	89326965	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 1
NM_002693.2(POLG):c.1399G>A (p.Ala467Thr)	POLG	15	89327201	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 1;Cerebellar ataxia infantile with progressive external ophthalmoplegia;Myoclonic epilepsy myopathy sensory ataxia;POLG-Related Spectrum Disorders;Progressive sclerosing poliodystrophy;Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis;not provided
NM_001151.3(SLC25A4):c.865G>A (p.Val289Met)	SLC25A4	4	185146939	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 2
NM_021830.4(TWNK):c.1075G>A (p.Ala359Thr)	TWNK	10	100989285	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 3
NM_021830.4(TWNK):c.1422G>A (p.Trp474Ter)	TWNK	10	100989822	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 3
NM_021830.4(TWNK):c.908G>A (p.Arg303Gln)	TWNK	10	100989118	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 3
NM_021830.4(TWNK):c.1120C>T (p.Arg374Trp)	TWNK	10	100989330	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 3
NM_007215.3(POLG2):c.1352G>A (p.Gly451Glu)	POLG2	17	64477929	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 4
NM_015713.4(RRM2B):c.979C>T (p.Arg327Ter)	RRM2B	8	102208210	Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 5;RRM2B-related mitochondrial disease;not provided
NM_001289123.1(TUBB4A):c.964G>A (p.Ala322Thr)	TUBB4A	19	6495688	Autosomal dominant torsion dystonia 4
NM_139343.2(BIN1):c.451G>A (p.Asp151Asn)	BIN1	2	127068992	Autosomal recessive centronuclear myopathy
NM_139343.2(BIN1):c.461G>A (p.Arg154Gln)	BIN1	2	127068982	Autosomal recessive centronuclear myopathy
NM_139343.2(BIN1):c.1713G>A (p.Trp571Ter)	BIN1	2	127048595	Autosomal recessive centronuclear myopathy
NM_000359.2(TGM1):c.428G>A (p.Arg143His)	TGM1	14	24261775	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.424C>T (p.Arg142Cys)	TGM1	14	24261779	Autosomal recessive congenital ichthyosis 1

NM_000359.2(TGM1):c.968G>A (p.Arg323Gln)	TGM1	14	24259720	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.425G>A (p.Arg142His)	TGM1	14	24261778	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.1147G>A (p.Val383Met)	TGM1	14	24259087	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.1166G>A (p.Arg389His)	TGM1	14	24258667	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.1175G>A (p.Gly392Asp)	TGM1	14	24258658	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.281G>A (p.Gly94Asp)	TGM1	14	24262072	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.919C>T (p.Arg307Trp)	TGM1	14	24259769	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.652G>A (p.Gly218Ser)	TGM1	14	24260555	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.1744C>T (p.Gln582Ter)	TGM1	14	24255155	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.376C>T (p.Arg126Cys)	TGM1	14	24261827	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.943C>T (p.Arg315Cys)	TGM1	14	24259745	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.944G>A (p.Arg315His)	TGM1	14	24259744	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.1075G>A (p.Val359Met)	TGM1	14	24259159	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.1187G>A (p.Arg396His)	TGM1	14	24258646	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.2278C>T (p.Arg760Ter)	TGM1	14	24249489	Autosomal recessive congenital ichthyosis 1
NM_000359.2(TGM1):c.832G>A (p.Gly278Arg)	TGM1	14	24259984	Autosomal recessive congenital ichthyosis 1;not provided
NM_000359.2(TGM1):c.857G>A (p.Arg286Gln)	TGM1	14	24259959	Autosomal recessive congenital ichthyosis 1;not provided
NM_001139.2(ALOX12B):c.340C>T (p.Arg114Trp)	ALOX12B	17	8086028	Autosomal recessive congenital ichthyosis 2
NM_001139.2(ALOX12B):c.1180G>A (p.Glu394Lys)	ALOX12B	17	8077085	Autosomal recessive congenital ichthyosis 2
NM_001139.2(ALOX12B):c.1207C>T (p.His403Tyr)	ALOX12B	17	8077058	Autosomal recessive congenital ichthyosis 2
NM_001139.2(ALOX12B):c.1642C>T (p.Arg548Trp)	ALOX12B	17	8075607	Autosomal recessive congenital ichthyosis 2;not provided
NM_021628.2(ALOXE3):c.418C>T (p.Arg140Ter)	ALOXE3	17	8115623	Autosomal recessive congenital ichthyosis 3
NM_021628.2(ALOXE3):c.700C>T (p.Arg234Ter)	ALOXE3	17	8112177	Autosomal recessive congenital ichthyosis 3;not provided
NM_173076.2(ABCA12):c.4142G>A (p.Gly1381Glu)	ABCA12	2	214986563	Autosomal recessive congenital ichthyosis 4A
NM_173076.2(ABCA12):c.4951G>A (p.Gly1651Ser)	ABCA12	2	214978830	Autosomal recessive congenital ichthyosis 4A
NM_173076.2(ABCA12):c.4541G>A (p.Arg1514His)	ABCA12	2	214982225	Autosomal recessive congenital ichthyosis 4A
NM_173076.2(ABCA12):c.4615G>A (p.Glu1539Lys)	ABCA12	2	214980608	Autosomal recessive congenital ichthyosis 4A
NM_173076.2(ABCA12):c.3535G>A (p.Gly1179Arg)	ABCA12	2	214990791	Autosomal recessive congenital ichthyosis 4B
NM_173076.2(ABCA12):c.6610C>T (p.Arg2204Ter)	ABCA12	2	214953891	Autosomal recessive congenital ichthyosis 4B;not provided
NM_173483.3(CYP4F22):c.728G>A (p.Arg243His)	CYP4F22	19	15540506	Autosomal recessive congenital ichthyosis 5
NM_173483.3(CYP4F22):c.1303C>T (p.His435Tyr)	CYP4F22	19	15549170	Autosomal recessive congenital ichthyosis 5;not provided
NM_001099287.1(NIPAL4):c.433C>T (p.Arg145Ter)	NIPAL4	5	157463303	Autosomal recessive congenital ichthyosis 6;not provided
NM_016938.4(EFEMP2):c.679C>T (p.Arg227Cys)	EFEMP2	11	65869905	Autosomal recessive cutis laxa type 1B
NM_016938.4(EFEMP2):c.169G>A (p.Glu57Lys)	EFEMP2	11	65871355	Autosomal recessive cutis laxa type 1B;Autosomal recessive cutis laxa type IA
NM_016938.4(EFEMP2):c.835C>T (p.Arg279Cys)	EFEMP2	11	65868522	Autosomal recessive cutis laxa type 1B;Autosomal recessive cutis laxa type IA
NM_016938.4(EFEMP2):c.1189G>A (p.Ala397Thr)	EFEMP2	11	65867061	Autosomal recessive cutis laxa type 1B;Autosomal recessive cutis laxa type IA
NM_016938.4(EFEMP2):c.376G>A (p.Glu126Lys)	EFEMP2	11	65870650	Autosomal recessive cutis laxa type 1B;Autosomal recessive cutis laxa type IA
NM_016938.4(EFEMP2):c.800G>A (p.Cys267Tyr)	EFEMP2	11	65868557	Autosomal recessive cutis laxa type 1B;Autosomal recessive cutis laxa type IA
NM_006907.3(PYCR1):c.797G>A (p.Arg266Gln)	PYCR1	17	81934326	Autosomal recessive cutis laxa type 2B
NM_006907.3(PYCR1):c.356G>A (p.Arg119His)	PYCR1	17	81935110	Autosomal recessive cutis laxa type 2B
NM_006907.3(PYCR1):c.752G>A (p.Arg251His)	PYCR1	17	81934371	Autosomal recessive cutis laxa type 3B
NM_006907.3(PYCR1):c.769G>A (p.Ala257Thr)	PYCR1	17	81934354	Autosomal recessive cutis laxa type 3B;not provided
NM_006907.3(PYCR1):c.743G>A (p.Gly248Glu)	PYCR1	17	81934380	Autosomal recessive cutis laxa type 3B;not provided
NM_181882.2(PRX):c.1102C>T (p.Arg368Ter)	PRX	19	40397250	Autosomal recessive Dejerine-Sottas syndrome
NM_000304.3(PMP22):c.469C>T (p.Arg157Trp)	PMP22	17	15230931	Autosomal recessive Dejerine-Sottas syndrome;Charcot-Marie-Tooth disease and deafness

NM_181882.2(PRX):c.2857C>T (p.Arg953Ter)	PRX	19	40395495	Autosomal recessive Dejerine-Sottas syndrome;Charcot-Marie-Tooth disease, demyelinating, type 4f;Charcot-Marie-Tooth disease, type IV;not provided
NM_022336.3(EDAR):c.51+1G>A	EDAR	2	108930963	Autosomal recessive hypohidrotic ectodermal dysplasia syndrome
NM_022336.3(EDAR):c.1124G>A (p.Arg375His)	EDAR	2	108897130	Autosomal recessive hypohidrotic ectodermal dysplasia syndrome
NM_022336.3(EDAR):c.803+1G>A	EDAR	2	108910459	Autosomal recessive hypohidrotic ectodermal dysplasia syndrome
NM_022336.3(EDAR):c.266G>A (p.Arg89His)	EDAR	2	108929288	Autosomal recessive hypohidrotic ectodermal dysplasia syndrome;Ectodermal dysplasia 10a, hypohidrotic/hair/tooth type, autosomal dominant
NM_080738.3(EDARADD):c.424G>A (p.Glu142Lys)	EDARADD	1	236482455	Autosomal recessive hypohidrotic ectodermal dysplasia syndrome;Ectodermal dysplasia 11b, hypohidrotic/hair/tooth type, autosomal recessive
NM_022336.3(EDAR):c.1144G>A (p.Gly382Ser)	EDAR	2	108897110	Autosomal recessive hypohidrotic ectodermal dysplasia syndrome;not provided
NM_001177317.1(SLC34A3):c.846G>A (p.Pro282=)	SLC34A3	9	137233722	Autosomal recessive hypophosphatemic bone disease
NM_001177317.1(SLC34A3):c.586G>A (p.Gly196Arg)	SLC34A3	9	137233234	Autosomal recessive hypophosphatemic bone disease
NM_001177317.1(SLC34A3):c.1402C>T (p.Arg468Trp)	SLC34A3	9	137236018	Autosomal recessive hypophosphatemic bone disease
NM_001177317.1(SLC34A3):c.756G>A (p.Gln252=)	SLC34A3	9	137233404	Autosomal recessive hypophosphatemic bone disease
NM_013296.4(GPSM2):c.977G>A (p.Trp326Ter)	GPSM2	1	108903149	Autosomal recessive non-syndromic sensorineural deafness type DFNB
NM_013296.4(GPSM2):c.379C>T (p.Arg127Ter)	GPSM2	1	108897592	Autosomal recessive non-syndromic sensorineural deafness type DFNB;Chudley-McCullough syndrome
NM_138694.3(PKHD1):c.4991C>T (p.Ser1664Phe)	PKHD1	6	52024819	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.9053C>T (p.Ser3018Phe)	PKHD1	6	51748563	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.8011C>T (p.Arg2671Ter)	PKHD1	6	51847871	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.1486C>T (p.Arg496Ter)	PKHD1	6	52058349	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.8303-1G>A	PKHD1	6	51791374	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.10444C>T (p.Arg3482Cys)	PKHD1	6	51659682	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.9319C>T (p.Arg3107Ter)	PKHD1	6	51748297	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.1480C>T (p.Arg494Ter)	PKHD1	6	52058355	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.2854G>A (p.Gly952Arg)	PKHD1	6	52043102	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.7194G>A (p.Trp2398Ter)	PKHD1	6	51885888	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.10219C>T (p.Gln3407Ter)	PKHD1	6	51659907	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.707+1G>A	PKHD1	6	52070405	Autosomal recessive polycystic kidney disease
NM_138694.3(PKHD1):c.107C>T (p.Thr36Met)	PKHD1	6	52083201	Autosomal recessive polycystic kidney disease;Colorectal cancer, protection against;Oligohydramnios;Periportal fibrosis;Polycystic kidney dysplasia;not provided
NM_138694.3(PKHD1):c.1602+1G>A	PKHD1	6	52056889	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.1694-1G>A	PKHD1	6	52055730	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.2341C>T (p.Arg781Ter)	PKHD1	6	52048558	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.2407+1G>A	PKHD1	6	52048491	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.2452C>T (p.Gln818Ter)	PKHD1	6	52046144	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.5236+1G>A	PKHD1	6	52024573	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.8824C>T (p.Arg2942Ter)	PKHD1	6	51753327	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.982C>T (p.Arg328Ter)	PKHD1	6	52062655	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.6499C>T (p.Gln2167Ter)	PKHD1	6	51909466	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.2725C>T (p.Arg909Ter)	PKHD1	6	52043721	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.370C>T (p.Arg124Ter)	PKHD1	6	52079920	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.4870C>T (p.Arg1624Trp)	PKHD1	6	52024940	Autosomal recessive polycystic kidney disease;not provided
NM_138694.3(PKHD1):c.2810G>A (p.Trp937Ter)	PKHD1	6	52043636	Autosomal recessive polycystic kidney disease;Polycystic kidney dysplasia;Renal cyst;Ventricular hypertrophy

NM_001098426.1(SMARCD2):c.1181+1G>A	SMARCD2	17	63833908	Autosomal recessive severe congenital neutropenia;Specific granule deficiency
NM_001844.4(COL2A1):c.2149G>A (p.Gly717Ser)	COL2A1	12	47982892	Avascular necrosis of the head of femur
NM_001844.4(COL2A1):c.4148C>T (p.Thr1383Met)	COL2A1	12	47974258	Avascular necrosis of the head of femur
NM_001844.4(COL2A1):c.3508G>A (p.Gly1170Ser)	COL2A1	12	47976052	Avascular necrosis of the head of femur;Coxa plana
NM_000358.2(TGFBI):c.371G>A (p.Arg124His)	TGFBI	5	136046407	Avellino corneal dystrophy
NM_153427.2(PITX2):c.399G>A (p.Trp133Ter)	PITX2	4	110618542	Axenfeld-Rieger syndrome type 1
NM_153427.2(PITX2):c.191C>T (p.Pro64Leu)	PITX2	4	110621225	Axenfeld-Rieger syndrome type 1
NM_001453.2(FOXC1):c.67C>T (p.Gln23Ter)	FOXC1	6	1610512	Axenfeld-Rieger syndrome type 3
NM_001453.2(FOXC1):c.358C>T (p.Gln120Ter)	FOXC1	6	1610803	Axenfeld-Rieger syndrome type 3
NM_001453.2(FOXC1):c.316C>T (p.Gln106Ter)	FOXC1	6	1610761	Axenfeld-Rieger syndrome type 3
NM_001453.2(FOXC1):c.256C>T (p.Leu86Phe)	FOXC1	6	1610701	Axenfeld-Rieger syndrome type 3;not provided
NM_000267.3(NF1):c.2041C>T (p.Arg681Ter)	NF1	17	31226474	Axillary freckling;Cafe-au-lait spot;Neurofibromatosis, type 1;Optic glioma;not provided
NM_005360.4(MAF):c.176C>T (p.Pro59Leu)	MAF	16	79599727	Ayme-gripp syndrome
NM_005360.4(MAF):c.161C>T (p.Ser54Leu)	MAF	16	79599742	Ayme-gripp syndrome
NM_005360.4(MAF):c.173C>T (p.Thr58Ile)	MAF	16	79599730	Ayme-gripp syndrome;not provided
NM_001039661.1(TIRAP):c.539C>T (p.Ser180Leu)	TIRAP	11	126292948	Bacteremia, resistance to;Invasive pneumococcal disease, protection against;Malaria, resistance to;Mycobacterium tuberculosis, protection against
NM_030632.2(ASXL3):c.1210C>T (p.Gln404Ter)	ASXL3	18	33738614	Bainbridge-Ropers syndrome
NM_030632.2(ASXL3):c.1396C>T (p.Gln466Ter)	ASXL3	18	33738800	Bainbridge-Ropers syndrome
NM_030632.2(ASXL3):c.3349C>T (p.Arg1117Ter)	ASXL3	18	33743197	Bainbridge-Ropers syndrome;not provided
NM_004260.3(RECQL4):c.3061C>T (p.Arg1021Trp)	RECQL4	8	144512319	Baller-Gerold syndrome
NM_004260.3(RECQL4):c.2269C>T (p.Gln757Ter)	RECQL4	8	144513412	Baller-Gerold syndrome;Rothmund-Thomson syndrome
NM_004473.3(FOXE1):c.194C>T (p.Ala65Val)	FOXE1	9	97854108	Bamforth syndrome
NM_004473.3(FOXE1):c.170G>A (p.Ser57Asn)	FOXE1	9	97854084	Bamforth syndrome
NM_004434.2(EML1):c.412C>T (p.Arg138Ter)	EML1	14	99878513	Band heterotopia of brain
NM_002538.3(OCLN):c.1037+5G>A	OCLN	5	69534844	Band-like calcification with simplified gyration and polymicrogyria
NM_000314.6(PTEN):c.697C>T (p.Arg233Ter)	PTEN	10	87957915	Bannayan-Riley-Ruvalcaba syndrome;Colorectal Neoplasms;Cowden syndrome 1;Glioblastoma;Hereditary cancer-predisposing syndrome;Macrocephaly/autism syndrome;Neoplasm of breast;Non-small cell lung cancer;PTEN hamartoma tumor syndrome;not provided
NM_000314.6(PTEN):c.388C>T (p.Arg130Ter)	PTEN	10	87933147	Bannayan-Riley-Ruvalcaba syndrome;Cowden syndrome 1;Hereditary cancer-predisposing syndrome;Macrocephaly/autism syndrome;Neoplasm of brain;Ovarian Neoplasms;PTEN hamartoma tumor syndrome;not provided
NM_000314.6(PTEN):c.1003C>T (p.Arg335Ter)	PTEN	10	87961095	Bannayan-Riley-Ruvalcaba syndrome;Cowden syndrome 1;Hereditary cancer-predisposing syndrome;Macrocephaly/autism syndrome;Neoplasm of brain;PTEN hamartoma tumor syndrome;Proteus-like syndrome;not provided
NM_000314.6(PTEN):c.640C>T (p.Gln214Ter)	PTEN	10	87957858	Bannayan-Riley-Ruvalcaba syndrome;Macrocephaly/autism syndrome
NM_000314.6(PTEN):c.511C>T (p.Gln171Ter)	PTEN	10	87952136	Bannayan-Riley-Ruvalcaba syndrome;PTEN hamartoma tumor syndrome;not provided
NM_001101.3(ACTB):c.349G>A (p.Glu117Lys)	ACTB	7	5529175	Baraitser-Winter syndrome 1
NM_001101.3(ACTB):c.193C>T (p.Leu65Phe)	ACTB	7	5529331	Baraitser-Winter syndrome 1
NM_001101.3(ACTB):c.209C>T (p.Pro70Leu)	ACTB	7	5529315	Baraitser-Winter syndrome 1
NM_001101.3(ACTB):c.359C>T (p.Thr120Ile)	ACTB	7	5529165	Baraitser-Winter syndrome 1
NM_001101.3(ACTB):c.446C>T (p.Thr149Ile)	ACTB	7	5528637	Baraitser-Winter syndrome 1
NM_001101.3(ACTB):c.625G>A (p.Val209Met)	ACTB	7	5528458	Baraitser-Winter syndrome 1
NM_001101.3(ACTB):c.1090G>A (p.Glu364Lys)	ACTB	7	5527786	Baraitser-Winter syndrome 1
NM_001101.3(ACTB):c.587G>A (p.Arg196His)	ACTB	7	5528496	Baraitser-Winter syndrome 1;not provided

NM_001101.3(ACTB):c.586C>T (p.Arg196Cys)	ACTB	7	5528497	Baraitser-Winter syndrome 1;not provided
NM_001101.3(ACTB):c.220G>A (p.Gly74Ser)	ACTB	7	5529304	Baraitser-Winter syndrome 1;not provided
NM_001614.3(ACTG1):c.464C>T (p.Ser155Phe)	ACTG1	17	81511526	Baraitser-Winter Syndrome 2;not provided
NM_001614.3(ACTG1):c.359C>T (p.Thr120Ile)	ACTG1	17	81511907	Baraitser-Winter Syndrome 2;not provided
NM_001614.3(ACTG1):c.404C>T (p.Ala135Val)	ACTG1	17	81511586	Baraitser-Winter Syndrome 2;not provided
NM_001614.3(ACTG1):c.760C>T (p.Arg254Trp)	ACTG1	17	81511230	Baraitser-Winter Syndrome 2;not provided
NM_001614.3(ACTG1):c.766C>T (p.Arg256Trp)	ACTG1	17	81511224	Baraitser-Winter Syndrome 2;not provided
NM_001002295.1(GATA3):c.829C>T (p.Arg277Ter)	GATA3	10	8064043	Barakat syndrome
NM_001002295.1(GATA3):c.1099C>T (p.Arg367Ter)	GATA3	10	8073787	Barakat syndrome
NM_001002295.1(GATA3):c.1025G>A (p.Cys342Tyr)	GATA3	10	8069573	Barakat syndrome
NM_152384.2(BBS5):c.413G>A (p.Arg138His)	BBS5	2	169492900	Bardet-Biedl syndrome
NM_024649.4(BBS1):c.1012C>T (p.Gln338Ter)	BBS1	11	66523784	Bardet-Biedl syndrome
NM_024649.4(BBS1):c.416G>A (p.Trp139Ter)	BBS1	11	66514662	Bardet-Biedl syndrome
NM_198428.2(BBS9):c.223C>T (p.Arg75Ter)	BBS9	7	33152811	Bardet-Biedl syndrome
NM_198428.2(BBS9):c.2115+1G>A	BBS9	7	33388145	Bardet-Biedl syndrome
NM_033028.4(BBS4):c.172C>T (p.Gln58Ter)	BBS4	15	72712259	Bardet-Biedl syndrome
NM_024649.4(BBS1):c.871C>T (p.Gln291Ter)	BBS1	11	66523496	Bardet-Biedl syndrome
NM_198428.2(BBS9):c.263+1G>A	BBS9	7	33152852	Bardet-Biedl syndrome
NM_024685.3(BBS10):c.1202G>A (p.Gly401Glu)	BBS10	12	76346783	Bardet-Biedl syndrome 10
NM_001178007.1(BBS12):c.1063C>T (p.Arg355Ter)	BBS12	4	122742955	Bardet-Biedl syndrome 12
NM_001178007.1(BBS12):c.1704G>A (p.Trp568Ter)	BBS12	4	122743596	Bardet-Biedl syndrome 12
NM_006642.4(SDCCAG8):c.740+356C>T	SDCCAG8	1	243305133	Bardet-Biedl syndrome 16
NM_001276378.1(LZTFL1):c.271C>T (p.Arg91Ter)	LZTFL1	3	45835591	Bardet-Biedl syndrome 17
NM_001177701.2(IFT27):c.299G>A (p.Cys100Tyr)	IFT27	22	36763972	Bardet-Biedl syndrome 19
NM_031885.3(BBS2):c.175C>T (p.Gln59Ter)	BBS2	16	56514623	Bardet-Biedl syndrome 2
NM_031885.3(BBS2):c.1864C>T (p.Arg622Ter)	BBS2	16	56497013	Bardet-Biedl syndrome 2;not provided
NM_177965.3(C8orf37):c.529C>T (p.Arg177Trp)	C8orf37	8	95247712	BARDET-BIEDL SYNDROME 21;Cone-rod dystrophy 16
NM_032146.5(ARL6):c.364C>T (p.Arg122Ter)	ARL6	3	97788004	Bardet-Biedl syndrome 3
NM_032146.5(ARL6):c.92C>T (p.Thr31Met)	ARL6	3	97768199	Bardet-Biedl syndrome 3
NM_033028.4(BBS4):c.883C>T (p.Arg295Ter)	BBS4	15	72731573	Bardet-Biedl syndrome 4
NM_152384.2(BBS5):c.214G>A (p.Gly72Ser)	BBS5	2	169487811	Bardet-Biedl syndrome 5
NM_170784.2(MKKS):c.155G>A (p.Gly52Asp)	MKKS	20	10413360	Bardet-Biedl syndrome 6
NM_144596.3(TTC8):c.624+1G>A	TTC8	14	88843851	Bardet-Biedl syndrome 8
NM_198428.2(BBS9):c.1792C>T (p.Arg598Ter)	BBS9	7	33383668	Bardet-Biedl syndrome 9
NM_198428.2(BBS9):c.421G>A (p.Gly141Arg)	BBS9	7	33177570	Bardet-Biedl syndrome 9
NM_198428.2(BBS9):c.1063C>T (p.Gln355Ter)	BBS9	7	33336487	Bardet-Biedl syndrome 9
NM_198428.2(BBS9):c.1759C>T (p.Arg587Ter)	BBS9	7	33367832	Bardet-Biedl syndrome 9
NM_198428.2(BBS9):c.1789+1G>A	BBS9	7	33367863	Bardet-Biedl syndrome 9;not provided
NM_024649.4(BBS1):c.432+1G>A	BBS1	11	66514679	Bardet-Biedl syndrome;Bardet-Biedl syndrome 1
NM_176824.2(BBS7):c.632C>T (p.Thr211Ile)	BBS7	4	121854790	Bardet-Biedl syndrome;Bardet-Biedl syndrome 1/7, digenic
NM_012210.3(TRIM32):c.388C>T (p.Pro130Ser)	TRIM32	9	116598130	Bardet-Biedl syndrome;Bardet-Biedl syndrome 11
NM_031885.3(BBS2):c.823C>T (p.Arg275Ter)	BBS2	16	56502790	Bardet-Biedl syndrome;Bardet-Biedl syndrome 2
NM_024685.3(BBS10):c.145C>T (p.Arg49Trp)	BBS10	12	76348214	Bardet-Biedl syndrome;inborn genetic diseases
NM_000246.3(CIITA):c.3234_3317del84	CIITA	16	10922491	Bare lymphocyte syndrome type 2, complementation group A
NM_000246.3(CIITA):c.2063G>A (p.Trp688Ter)	CIITA	16	10907555	Bare lymphocyte syndrome type 2, complementation group A

NM_000449.3(RFX5):c.446G>A (p.Arg149Gln)	RFX5	1	151344444	Bare lymphocyte syndrome type 2, complementation group E
NM_000538.3(RFXAP):c.163C>T (p.Gln55Ter)	RFXAP	13	36819520	Bare Lymphocyte Syndrome, Type II, Complementation Group D
NM_000085.4(CLCNKB):c.371C>T (p.Pro124Leu)	CLCNKB	1	16047917	Bartter syndrome type 3
NM_000085.4(CLCNKB):c.610G>A (p.Ala204Thr)	CLCNKB	1	16048537	Bartter syndrome type 3
NM_057176.2(BSND):c.22C>T (p.Arg8Trp)	BSND	1	54999208	Bartter syndrome type 4
NM_057176.2(BSND):c.3G>A (p.Met1Ile)	BSND	1	54999189	Bartter syndrome type 4
NM_057176.2(BSND):c.28G>A (p.Gly10Ser)	BSND	1	54999214	Bartter syndrome type 4
NM_057176.2(BSND):c.139G>A (p.Gly47Arg)	BSND	1	54999325	Bartter syndrome type 4
NM_000338.2(SLC12A1):c.1942G>A (p.Asp648Asn)	SLC12A1	15	48251770	Bartter syndrome, type 1, antenatal
NM_000338.2(SLC12A1):c.1875G>A (p.Trp625Ter)	SLC12A1	15	48251703	Bartter syndrome, type 1, antenatal
NM_153767.3(KCNJ1):c.584C>T (p.Ala195Val)	KCNJ1	11	128839660	Bartter syndrome, type 2, antenatal
NM_153767.3(KCNJ1):c.535G>A (p.Ala179Thr)	KCNJ1	11	128839709	Bartter syndrome, type 2, antenatal
NM_153767.3(KCNJ1):c.443G>A (p.Gly148Glu)	KCNJ1	11	128839801	Bartter syndrome, type 2, antenatal
NM_000085.4(CLCNKB):c.1830G>A (p.Trp610Ter)	CLCNKB	1	16035508	Bartter syndrome, type 3, with hypocalciuria;not provided
NM_004070.3(CLCNKA):c.778C>T (p.Gln260Ter)	CLCNKA	1	16027432	Bartter syndrome, type 4b
NM_177433.2(MAGED2):c.1336C>T (p.Arg446Cys)	MAGED2	X	54814725	Bartter syndrome, type 5, antenatal, transient
NM_003738.4(PTCH2):c.3357+5C>T	PTCH2	1	44823064	Basal cell carcinoma, somatic
NM_005631.4(SMO):c.1685G>A (p.Arg562Gln)	SMO	7	129210997	Basal cell carcinoma, somatic
NM_002609.3(PDGFRB):c.2959C>T (p.Arg987Trp)	PDGFRB	5	150117796	Basal ganglia calcification, idiopathic, 4
NM_004736.3(XPR1):c.407G>A (p.Ser136Asn)	XPR1	1	180803571	Basal ganglia calcification, idiopathic, 6
NM_000186.3(CFH):c.1222C>T (p.Gln408Ter)	CFH	1	196690125	Basal laminar drusen
NM_032415.5(CARD11):c.367G>A (p.Gly123Ser)	CARD11	7	2944529	B-cell expansion with NFKB and T-cell anergy
NM_032415.5(CARD11):c.368G>A (p.Gly123Asp)	CARD11	7	2944528	B-cell expansion with NFKB and T-cell anergy
NM_032415.5(CARD11):c.146G>A (p.Cys49Tyr)	CARD11	7	2947649	B-cell expansion with NFKB and T-cell anergy
NM_004328.4(BCS1L):c.166C>T (p.Arg56Ter)	BCS1L	2	218661153	BCS1L-Related Disorders;Mitochondrial complex III deficiency;not provided
NM_002282.3(KRT83):c.1219G>A (p.Glu407Lys)	KRT82;KRT83	12	52315936	Beaded hair;not provided
NM_002281.3(KRT81):c.1237G>A (p.Glu413Lys)	KRT81	12	52287112	Beaded hair;not provided
NM_002281.3(KRT81):c.1204G>A (p.Glu402Lys)	KRT81	12	52287145	Beaded hair;not provided
NM_001320198.1(KRT86):c.1237G>A (p.Glu413Lys)	KRT86	12	52306270	Beaded hair;not provided
NM_001320198.1(KRT86):c.1204G>A (p.Glu402Lys)	KRT86	12	52306237	Beaded hair;not provided
NM_024339.4(THOC6):c.136G>A (p.Gly46Arg)	THOC6	16	3023804	Beaulieu-Boycott-Innes syndrome
NM_004006.2(DMD):c.3413G>A (p.Trp1138Ter)	DMD	X	32463458	Becker muscular dystrophy
NM_004006.2(DMD):c.358-1G>A	DMD	X	32816641	Becker muscular dystrophy
NM_004006.2(DMD):c.8944C>T (p.Arg2982Ter)	DMD	X	31444621	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.10108C>T (p.Arg3370Ter)	DMD	X	31178784	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.5899C>T (p.Arg1967Ter)	DMD	X	32342123	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.6373C>T (p.Gln2125Ter)	DMD	X	32216981	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.9568C>T (p.Arg3190Ter)	DMD	X	31206663	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.8713C>T (p.Arg2905Ter)	DMD	X	31478330	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.10033C>T (p.Arg3345Ter)	DMD	X	31180423	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.10086+1G>A	DMD	X	31180369	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004019.2(DMD):c.1020G>A (p.Thr340=)	DMD	X	31178668	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.1261C>T (p.Gln421Ter)	DMD	X	32644202	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.1465C>T (p.Gln489Ter)	DMD	X	32614320	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.1615C>T (p.Arg539Ter)	DMD	X	32573834	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy

NM_004006.2(DMD):c.1990C>T (p.Gln664Ter)	DMD	X	32565704	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.2032C>T (p.Gln678Ter)	DMD	X	32545295	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.2332C>T (p.Gln778Ter)	DMD	X	32501803	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.2419C>T (p.Gln807Ter)	DMD	X	32491480	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.2650C>T (p.Gln884Ter)	DMD	X	32485072	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.2804-1G>A	DMD	X	32472310	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.3151C>T (p.Arg1051Ter)	DMD	X	32468509	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.3276+1G>A	DMD	X	32464585	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.3295C>T (p.Gln1099Ter)	DMD	X	32463576	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.3366G>A (p.Trp112Ter)	DMD	X	32823316	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.3432+1G>A	DMD	X	32463438	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.3580C>T (p.Gln1194Ter)	DMD	X	32454685	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.4117C>T (p.Gln1373Ter)	DMD	X	32411868	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.5287C>T (p.Arg1763Ter)	DMD	X	32362826	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.5530C>T (p.Arg1844Ter)	DMD	X	32345999	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.649+1G>A	DMD	X	32809492	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.6906G>A (p.Trp2302Ter)	DMD	X	31929602	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.7189C>T (p.Gln2397Ter)	DMD	X	31836729	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.7309+1G>A	DMD	X	31819974	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.7657C>T (p.Arg2553Ter)	DMD	X	31729634	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.7682G>A (p.Trp2561Ter)	DMD	X	31679565	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.7683G>A (p.Trp2561Ter)	DMD	X	31679564	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.7894C>T (p.Gln2632Ter)	DMD	X	31658123	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.8608C>T (p.Arg2870Ter)	DMD	X	31479043	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.9361+1G>A	DMD	X	31223046	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.9564-1G>A	DMD	X	31206668	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.2956C>T (p.Gln986Ter)	DMD	X	32468704	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.883C>T (p.Arg295Ter)	DMD	X	32697947	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.8656C>T (p.Gln2886Ter)	DMD	X	31478995	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.31+36947G>A	DMD	X	33174335	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.10279C>T (p.Gln3427Ter)	DMD	X	31173588	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.433C>T (p.Arg145Ter)	DMD	X	32816565	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy;not provided
NM_004006.2(DMD):c.9G>A (p.Trp3Ter)	DMD	X	33211304	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy;not provided
NM_004006.2(DMD):c.10171C>T (p.Arg3391Ter)	DMD	X	31178721	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy;not provided
NM_004006.2(DMD):c.583C>T (p.Arg195Ter)	DMD	X	32809559	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy;not provided
NM_004006.2(DMD):c.9337C>T (p.Arg3113Ter)	DMD	X	31223071	Becker muscular dystrophy;Dilated cardiomyopathy 3B;Duchenne muscular dystrophy;not provided
NM_004006.2(DMD):c.1812+1G>A	DMD	X	32573529	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.1093C>T (p.Gln365Ter)	DMD	X	32645020	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.1704+1G>A	DMD	X	32573744	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.1912C>T (p.Gln638Ter)	DMD	X	32565782	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.133C>T (p.Gln45Ter)	DMD	X	32849781	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.5868G>A (p.Trp1956Ter)	DMD	X	32342154	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.565C>T (p.Gln189Ter)	DMD	X	32809577	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.8038C>T (p.Arg2680Ter)	DMD	X	31627852	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.5089C>T (p.Gln1697Ter)	DMD	X	32364647	Becker muscular dystrophy;Duchenne muscular dystrophy

NM_004006.2(DMD):c.2512C>T (p.Gln838Ter)	DMD	X	32491387	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.10477C>T (p.Gln3493Ter)	DMD	X	31169519	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.93+1G>A	DMD	X	33020138	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.4174C>T (p.Gln1392Ter)	DMD	X	32411811	Becker muscular dystrophy;Duchenne muscular dystrophy
NM_004006.2(DMD):c.3940C>T (p.Arg1314Ter)	DMD	X	32438372	Becker muscular dystrophy;Duchenne muscular dystrophy;not provided
NM_000076.2(CDKN1C):c.139C>T (p.Gln47Ter)	CDKN1C	11	2885351	Beckwith-Wiedemann syndrome
NM_000076.2(CDKN1C):c.694C>T (p.Gln232Ter)	CDKN1C	11	2884796	Beckwith-Wiedemann syndrome
NM_022455.4(NSD1):c.6487C>T (p.Gln2163Ter)	NSD1	5	177293855	Beckwith-Wiedemann syndrome
NM_022455.4(NSD1):c.5581C>T (p.Arg1861Ter)	NSD1	5	177273743	Beckwith-Wiedemann syndrome;not provided
NM_022455.4(NSD1):c.2362C>T (p.Arg788Ter)	NSD1	5	177210761	Beckwith-Wiedemann syndrome;not provided
NM_022455.4(NSD1):c.4411C>T (p.Arg1471Ter)	NSD1	5	177246710	Beckwith-Wiedemann syndrome;Sotos syndrome 1
NM_022455.4(NSD1):c.1810C>T (p.Arg604Ter)	NSD1	5	177210209	Beckwith-Wiedemann syndrome;Sotos syndrome 1;not provided
NM_022455.4(NSD1):c.6349C>T (p.Arg2117Ter)	NSD1	5	177292044	Beckwith-Wiedemann syndrome;Sotos syndrome 1;not provided
NM_022162.2(NOD2):c.2446G>A	NOD2	16	50712357	Behcet's syndrome
NM_001065.3(TNFRSF1A):c.463C>T	TNFRSF1A	12	6333376	Behcet's syndrome
NM_022162.2(NOD2):c.1045C>T (p.Leu349Phe)	NOD2	16	50710956	Behcet's syndrome
NM_001282225.1(ADA2):c.145C>T (p.Arg49Trp)	ADA2	22	17209533	Behcet's syndrome
NM_001282225.1(ADA2):c.927G>A (p.Met309Ile)	ADA2	22	17189987	Behcet's syndrome
NM_001282225.1(ADA2):c.740C>T (p.Ala247Val)	ADA2	22	17203576	Behcet's syndrome
NM_000092.4(COL4A4):c.2690G>A (p.Gly897Glu)	COL4A4	2	227055971	Benign familial hematuria
NM_000091.4(COL4A3):c.3044G>A (p.Gly1015Glu)	COL4A3	2	227290062	Benign familial hematuria
NM_000092.4(COL4A4):c.2986G>A (p.Gly996Arg)	COL4A4	2	227051141	Benign familial hematuria
NM_000091.4(COL4A3):c.3410G>A (p.Gly1137Asp)	COL4A3	2	227294562	Benign familial hematuria
NM_172107.3(KCNQ2):c.916G>A (p.Ala306Thr)	KCNQ2	20	63439609	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.1118+1G>A	KCNQ2	20	63433808	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.1288C>T (p.Pro430Ser)	KCNQ2	20	63419632	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.1525+1G>A	KCNQ2	20	63414902	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.587C>T (p.Ala196Val)	KCNQ2	20	63444762	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.727C>T (p.Leu243Phe)	KCNQ2	20	63442495	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.967C>T (p.Gln323Ter)	KCNQ2	20	63438681	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.296+1G>A	KCNQ2	20	63472167	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.319C>T (p.Leu107Phe)	KCNQ2	20	63446815	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.1783C>T (p.Arg595Trp)	KCNQ2	20	63408517	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.1247+1G>A	KCNQ2	20	63424176	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.1073C>T (p.Ser358Phe)	KCNQ2	20	63433854	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.1051C>T (p.Leu351Phe)	KCNQ2	20	63433876	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.476G>A (p.Gly159Glu)	KCNQ2	20	63445276	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.475G>A (p.Gly159Arg)	KCNQ2	20	63445277	Benign familial neonatal seizures 1
NM_172107.3(KCNQ2):c.869G>A (p.Gly290Asp)	KCNQ2	20	63439656	Benign familial neonatal seizures 1;Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.1657C>T (p.Arg553Trp)	KCNQ2	20	63413556	Benign familial neonatal seizures 1;Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.793G>A (p.Ala265Thr)	KCNQ2	20	63442429	Benign familial neonatal seizures 1;Early infantile epileptic encephalopathy 7;Seizures;not provided
NM_172107.3(KCNQ2):c.1342C>T (p.Arg448Ter)	KCNQ2	20	63415086	Benign familial neonatal seizures 1;Early infantile epileptic encephalopathy;not provided
NM_172107.3(KCNQ2):c.1742G>A (p.Arg581Gln)	KCNQ2	20	63413471	Benign familial neonatal seizures 1;Epileptic encephalopathy;not provided
NM_172107.3(KCNQ2):c.640C>T (p.Arg214Trp)	KCNQ2	20	63444709	Benign familial neonatal seizures 1;not provided
NM_172107.3(KCNQ2):c.1658G>A (p.Arg553Gln)	KCNQ2	20	63413555	Benign familial neonatal seizures 1;not provided

NM_172107.3(KCNQ2):c.1741C>T (p.Arg581Ter)	KCNQ2	20	63413472	Benign familial neonatal seizures 1;not provided
NM_172107.3(KCNQ2):c.365C>T (p.Ser122Leu)	KCNQ2	20	63446769	Benign familial neonatal seizures 1;not provided
NM_172107.3(KCNQ2):c.807G>A (p.Trp269Ter)	KCNQ2	20	63442415	Benign familial neonatal seizures 1;not provided
NM_172107.3(KCNQ2):c.998G>A (p.Arg333Gln)	KCNQ2	20	63438650	Benign familial neonatal seizures 1;not provided
NM_172107.3(KCNQ2):c.1639C>T (p.Arg547Trp)	KCNQ2	20	63413574	Benign familial neonatal seizures 1;not provided
NM_172107.3(KCNQ2):c.1631+1G>A	KCNQ2	20	63414087	Benign familial neonatal seizures 1;not provided
NM_172107.3(KCNQ2):c.619C>T (p.Arg207Trp)	KCNQ2	20	63444730	Benign familial neonatal seizures 1;Seizures, benign familial neonatal, 1, and/or myokymia;not provided
NM_004519.3(KCNQ3):c.895G>A (p.Glu299Lys)	KCNQ3	8	132175491	Benign familial neonatal seizures 2
NM_004519.3(KCNQ3):c.688C>T (p.Arg230Cys)	KCNQ3	8	132180246	Benign familial neonatal seizures 2;not provided
NM_004519.3(KCNQ3):c.988C>T (p.Arg330Cys)	KCNQ3	8	132174295	Benign familial neonatal seizures;Benign familial neonatal seizures 2
NM_001040142.1(SCN2A):c.562C>T (p.Arg188Trp)	SCN2A	2	165308751	Benign familial neonatal-infantile seizures
NM_001040142.1(SCN2A):c.3988C>T (p.Leu1330Phe)	SCN2A	2	165374700	Benign familial neonatal-infantile seizures
NM_001040142.1(SCN2A):c.668G>A (p.Arg223Gln)	SCN2A	2	165309414	Benign familial neonatal-infantile seizures
NM_021007.2(SCN2A):c.4876C>T (p.Arg1626Ter)	SCN2A	2	165388682	Benign familial neonatal-infantile seizures;Early infantile epileptic encephalopathy 11
NM_021007.2(SCN2A):c.788C>T (p.Ala263Val)	SCN2A	2	165310413	Benign familial neonatal-infantile seizures;Early infantile epileptic encephalopathy 11;Epileptic encephalopathy;not provided
NM_021007.2(SCN2A):c.5645G>A (p.Arg1882Gln)	SCN2A	2	165389451	Benign familial neonatal-infantile seizures;Early infantile epileptic encephalopathy 11;Epileptic encephalopathy;not provided
NM_021007.2(SCN2A):c.3631G>A (p.Glu1211Lys)	SCN2A	2	165367327	Benign familial neonatal-infantile seizures;Early infantile epileptic encephalopathy 11;not provided
NM_021007.2(SCN2A):c.2674G>A (p.Val892Ile)	SCN2A	2	165344666	Benign familial neonatal-infantile seizures;not provided
NM_021007.2(SCN2A):c.3956G>A (p.Arg1319Gln)	SCN2A	2	165373331	Benign familial neonatal-infantile seizures;not provided
NM_001079668.2(NKX2-1):c.745C>T (p.Gln249Ter)	NKX2-1	14	36517739	Benign hereditary chorea
NM_003742.2(ABCB11):c.150+1G>A	ABCB11	2	169014302	Benign recurrent intrahepatic cholestasis 2
NM_003742.2(ABCB11):c.908+1G>A	ABCB11	2	168990800	Benign recurrent intrahepatic cholestasis 2;Progressive familial intrahepatic cholestasis 2
NM_003742.2(ABCB11):c.3692G>A (p.Arg1231Gln)	ABCB11	2	168924730	Benign recurrent intrahepatic cholestasis 2;Progressive familial intrahepatic cholestasis 2
NM_003742.2(ABCB11):c.22C>T (p.Arg8Ter)	ABCB11	2	169018104	Benign recurrent intrahepatic cholestasis 2;Progressive familial intrahepatic cholestasis 2
NM_003742.2(ABCB11):c.3457C>T (p.Arg1153Cys)	ABCB11	2	168927317	Benign recurrent intrahepatic cholestasis 2;Progressive familial intrahepatic cholestasis 2
NM_003742.2(ABCB11):c.2380C>T (p.Gln794Ter)	ABCB11	2	168944925	Benign recurrent intrahepatic cholestasis 2;Progressive familial intrahepatic cholestasis 2
NM_170707.3(LMNA):c.1072G>A (p.Glu358Lys)	LMNA	1	156136036	Benign scapuloperoneal muscular dystrophy with cardiomyopathy;Charcot-Marie-Tooth disease, type 2;Congenital muscular dystrophy, LMNA-related;Dilated cardiomyopathy 1A;Familial partial lipodystrophy 2;Limb-girdle muscular dystrophy, type 1B;Mandibuloacral dysostosis;not provided
NM_170707.3(LMNA):c.1357C>T (p.Arg453Trp)	LMNA	1	156136413	Benign scapuloperoneal muscular dystrophy with cardiomyopathy;Charcot-Marie-Tooth disease, type 2;Limb-girdle muscular dystrophy, type 1B;not provided
NM_170707.3(LMNA):c.745C>T (p.Arg249Trp)	LMNA	1	156134910	Benign scapuloperoneal muscular dystrophy with cardiomyopathy;Congenital muscular dystrophy, LMNA-related;not provided
NM_005572.3(LMNA):c.674G>A (p.Arg225Gln)	LMNA	1	156134839	Benign scapuloperoneal muscular dystrophy with cardiomyopathy;Emery-Dreifuss muscular dystrophy 3, autosomal recessive
NM_005572.3(LMNA):c.664C>T (p.His222Tyr)	LMNA	1	156134829	Benign scapuloperoneal muscular dystrophy with cardiomyopathy;Emery-Dreifuss muscular dystrophy 3, autosomal recessive;not provided
NM_170707.3(LMNA):c.746G>A (p.Arg249Gln)	LMNA	1	156134911	Benign scapuloperoneal muscular dystrophy with cardiomyopathy;not provided
NM_170707.3(LMNA):c.16C>T (p.Gln6Ter)	LMNA	1	156114934	Benign scapuloperoneal muscular dystrophy with cardiomyopathy;Primary dilated cardiomyopathy;not provided
NM_000173.6(GP1BA):c.1077G>A (p.Trp359Ter)	GP1BA	17	4933681	Bernard-Soulier syndrome, type A1
NM_000173.6(GP1BA):c.1620G>A (p.Trp540Ter)	GP1BA	17	4934224	Bernard-Soulier syndrome, type A1

NM_000173.6(GP1BA):c.515C>T (p.Ala172Val)	GP1BA	17	4933119	Bernard-Soulier syndrome, type A1;Bernard-Soulier syndrome, type A2, autosomal dominant
NM_000173.6(GP1BA):c.217C>T (p.Leu73Phe)	GP1BA	17	4932821	Bernard-Soulier syndrome, type A2, autosomal dominant
NM_000407.4(GP1BB):c.137G>A (p.Trp46Ter)	GP1BB	22	19723980	Bernard-Soulier syndrome, type B
NM_004183.3(BEST1):c.598C>T (p.Arg200Ter)	BEST1	11	61956960	Bestrophinopathy, autosomal recessive
NM_004183.3(BEST1):c.949G>A (p.Val317Met)	BEST1	11	61959892	Bestrophinopathy, autosomal recessive
NM_000518.4(HBB):c.-29G>A	HBB	11	5227050	beta Thalassemia
NM_000518.4(HBB):c.-137C>T	HBB	11	5227158	Beta thalassemia intermedia;beta Thalassemia
NM_000518.4(HBB):c.-50-88C>T	HBB	11	5227159	Beta thalassemia intermedia;Beta-plus-thalassemia;beta Thalassemia
NM_000518.4(HBB):c.-140C>T	HBB	11	5227161	Beta thalassemia intermedia;Beta-plus-thalassemia;beta Thalassemia
NM_000518.4(HBB):c.316-197C>T	HBB	11	5225923	Beta thalassemia major;beta Thalassemia;beta^0^ Thalassemia
NM_000518.4(HBB):c.93-21G>A	HBB	11	5226820	Beta thalassemia major;Beta-plus-thalassemia;beta Thalassemia
NM_000518.4(HBB):c.114G>A (p.Trp38Ter)	HBB	11	5226778	beta Thalassemia;beta^0^ Thalassemia
NM_000518.4(HBB):c.118C>T (p.Gln40Ter)	HBB	11	5226774	beta Thalassemia;beta^0^ Thalassemia;not provided
NM_000518.4(HBB):c.92+1G>A	HBB	11	5226929	beta Thalassemia;beta^0^ Thalassemia;not provided
NM_000518.4(HBB):c.315+1G>A	HBB	11	5226576	beta Thalassemia;beta^0^ Thalassemia;not provided
NM_000518.4(HBB):c.47G>A (p.Trp16Ter)	HBB	11	5226975	beta^0^ Thalassemia
NM_000518.4(HBB):c.93-1G>A	HBB	11	5226800	beta^0^ Thalassemia
NM_000518.4(HBB):c.3G>A (p.Met1Ile)	HBB	11	5227019	beta^0^ Thalassemia
NM_005908.3(MANBA):c.1276C>T (p.Gln426Ter)	MANBA	4	102669004	Beta-D-mannosidosis
NM_014362.3(HIBCH):c.950G>A (p.Gly317Glu)	HIBCH	2	190213017	Beta-hydroxyisobutyryl-CoA deacylase deficiency
NM_014362.3(HIBCH):c.196C>T (p.Arg66Trp)	HIBCH	2	190296836	Beta-hydroxyisobutyryl-CoA deacylase deficiency
NM_014362.3(HIBCH):c.1033G>A (p.Gly345Ser)	HIBCH	2	190208892	Beta-hydroxyisobutyryl-CoA deacylase deficiency
NM_000518.4(HBB):c.92+5G>A	HBB	11	5226925	Beta-plus-thalassemia;beta Thalassemia
NM_000518.4(HBB):c.-50-101C>T	HBB	11	5227172	Beta-plus-thalassemia;beta Thalassemia
NM_000518.4(HBB):c.-50-92C>T	HBB	11	5227163	Beta-plus-thalassemia;beta Thalassemia
NM_000518.4(HBB):c.79G>A (p.Glu27Lys)	HBB	11	5226943	Beta-plus-thalassemia;Hb SS disease;Hemoglobin E;Hemoglobin E/beta thalassemia disease;Malaria, resistance to
NM_000232.4(SGCB):c.341C>T (p.Ser114Phe)	SGCB	4	52029766	Beta-sarcoglycanopathy;Limb-girdle muscular dystrophy, type 2E;not provided
NM_001849.3(COL6A2):c.811G>A (p.Gly271Ser)	COL6A2	21	46115881	Bethlem myopathy 1
NM_001849.3(COL6A2):c.1861G>A (p.Asp621Asn)	COL6A2	21	46125509	Bethlem myopathy 1
NM_001848.2(COL6A1):c.1022G>A (p.Gly341Asp)	COL6A1	21	45990792	Bethlem myopathy 1
NM_001849.3(COL6A2):c.2611G>A (p.Asp871Asn)	COL6A2	21	46132103	Bethlem myopathy 1
NM_001849.3(COL6A2):c.901G>A (p.Gly301Ser)	COL6A2	21	46116377	Bethlem myopathy 1
NM_001849.3(COL6A2):c.1053+1G>A	COL6A2	21	46117454	Bethlem myopathy 1
NM_001849.3(COL6A2):c.1096C>T (p.Arg366Ter)	COL6A2	21	46117916	Bethlem myopathy 1;not provided
NM_004369.3(COL6A3):c.6239G>A (p.Gly2080Asp)	COL6A3	2	237360131	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001849.3(COL6A2):c.801+1G>A	COL6A2	21	46114074	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001849.3(COL6A2):c.812G>A (p.Gly271Asp)	COL6A2	21	46115882	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.931-1G>A	COL6A1	21	45990257	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.1611+1G>A	COL6A1	21	45998434	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.930+1G>A	COL6A1	21	45989779	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001849.3(COL6A2):c.785G>A (p.Gly262Asp)	COL6A2	21	46114057	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.1003-1G>A	COL6A1	21	45990772	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.1002+1G>A	COL6A1	21	45990423	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.932G>A (p.Gly311Asp)	COL6A1	21	45990259	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1

NM_001848.2(COL6A1):c.1003_1056del54	COL6A1	21	45990827	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;not provided
NM_001848.2(COL6A1):c.868G>A (p.Gly290Arg)	COL6A1	21	45989617	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;not provided
NM_001849.3(COL6A2):c.1522-1G>A	COL6A2	21	46122107	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;not provided
NM_004369.3(COL6A3):c.175C>T (p.Arg59Ter)	COL6A3	2	237395121	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;not provided
NM_004369.3(COL6A3):c.6210+1G>A	COL6A3	2	237361120	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;not provided
NM_004369.3(COL6A3):c.6282+1G>A	COL6A3	2	237360087	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;not provided
NM_001848.2(COL6A1):c.850G>A (p.Gly284Arg)	COL6A1	21	45989129	Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;Ullrich congenital muscular dystrophy 1, autosomal dominant;not provided
NM_004370.5(COL12A1):c.8357G>A (p.Gly2786Asp)	COL12A1	6	75102655	Bethlem myopathy 2
NM_207352.3(CYP4V2):c.1523G>A (p.Arg508His)	CYP4V2	4	186210586	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.181G>A (p.Gly61Ser)	CYP4V2	4	186192004	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.1020G>A (p.Trp340Ter)	CYP4V2	4	186205232	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.1169G>A (p.Arg390His)	CYP4V2	4	186208943	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.1187C>T (p.Pro396Leu)	CYP4V2	4	186208961	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.1199G>A (p.Arg400His)	CYP4V2	4	186208973	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.1348C>T (p.Gln450Ter)	CYP4V2	4	186209215	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.1526C>T (p.Pro509Leu)	CYP4V2	4	186210589	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.253C>T (p.Arg85Cys)	CYP4V2	4	186194538	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.283G>A (p.Gly95Arg)	CYP4V2	4	186194568	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.327+1G>A	CYP4V2	4	186194613	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.958C>T (p.Arg320Ter)	CYP4V2	4	186201313	Bietti crystalline corneoretinal dystrophy
NM_207352.3(CYP4V2):c.974C>T (p.Thr325Ile)	CYP4V2	4	186201329	Bietti crystalline corneoretinal dystrophy
NM_144966.5(FREM1):c.1945C>T (p.Arg649Trp)	FREM1	9	14824929	Bifid nose with or without anorectal and renal anomalies;not provided
NM_144966.5(FREM1):c.4318G>A (p.Gly1440Ser)	FREM1	9	14784494	Bifid nose with or without anorectal and renal anomalies;not provided
NM_000414.3(HSD17B4):c.46G>A (p.Gly16Ser)	HSD17B4	5	119452621	Bifunctional peroxisomal enzyme deficiency;not provided
NM_015160.2(PMPCA):c.1066G>A (p.Gly356Ser)	PMPCA	9	136418630	Bilateral ptosis;Blindness;Chronic lactic acidosis;Diffuse cerebellar atrophy;Failure to thrive;Global brain atrophy;Hypertrophic cardiomyopathy;Hypoventilation;Infantile muscular hypotonia;Normal pressure hydrocephalus;Optic atrophy;Restrictive external ophthalmoplegia;Severe global developmental delay
NM_015160.2(PMPCA):c.1129G>A (p.Ala377Thr)	PMPCA	9	136418847	Bilateral ptosis;Blindness;Chronic lactic acidosis;Diffuse cerebellar atrophy;Failure to thrive;Global brain atrophy;Hypertrophic cardiomyopathy;Hypoventilation;Infantile muscular hypotonia;Normal pressure hydrocephalus;Optic atrophy;Restrictive external ophthalmoplegia;Severe global developmental delay;Spinocerebellar ataxia, autosomal recessive 2
NM_025193.3(HSD3B7):c.439G>A (p.Glu147Lys)	HSD3B7	16	30986612	Bile acid synthesis defect, congenital, 1
NM_005989.3(AKR1D1):c.316C>T (p.Leu106Phe)	AKR1D1	7	138091822	Bile acid synthesis defect, congenital, 2
NM_005989.3(AKR1D1):c.781C>T (p.Arg261Cys)	AKR1D1	7	138107506	Bile acid synthesis defect, congenital, 2
NM_004820.4(CYP7B1):c.1162C>T (p.Arg388Ter)	CYP7B1	8	64604753	Bile acid synthesis defect, congenital, 3;Spastic paraplegia 5A
NM_003500.3(ACOX2):c.673C>T (p.Arg225Trp)	ACOX2	3	58531723	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 6
NM_000060.4(BTD):c.100G>A (p.Gly34Ser)	BTD	3	15635479	Biotinidase deficiency
NM_000060.4(BTD):c.235C>T (p.Arg79Cys)	BTD	3	15635614	Biotinidase deficiency
NM_000060.4(BTD):c.1157G>A (p.Trp386Ter)	BTD	3	15645013	Biotinidase deficiency
NM_000060.4(BTD):c.190G>A (p.Glu64Lys)	BTD	3	15635569	Biotinidase deficiency
NM_000060.4(BTD):c.245C>T (p.Ala82Val)	BTD	3	15635624	Biotinidase deficiency
NM_000060.4(BTD):c.262C>T (p.Gln88Ter)	BTD	3	15635641	Biotinidase deficiency
NM_000060.4(BTD):c.298G>A (p.Ala100Thr)	BTD	3	15635677	Biotinidase deficiency

NM_000060.4(BTD):c.334G>A (p.Glu112Lys)	BTD	3	15641932	Biotinidase deficiency
NM_000060.4(BTD):c.443G>A (p.Arg148His)	BTD	3	15642041	Biotinidase deficiency
NM_000060.4(BTD):c.466C>T (p.Gln156Ter)	BTD	3	15644322	Biotinidase deficiency
NM_000060.4(BTD):c.485C>T (p.Ala162Val)	BTD	3	15644341	Biotinidase deficiency
NM_000060.4(BTD):c.595G>A (p.Val199Met)	BTD	3	15644451	Biotinidase deficiency
NM_000060.4(BTD):c.631C>T (p.Arg211Cys)	BTD	3	15644487	Biotinidase deficiency
NM_000060.4(BTD):c.643C>T (p.Leu215Phe)	BTD	3	15644499	Biotinidase deficiency
NM_000060.4(BTD):c.709G>A (p.Ala237Thr)	BTD	3	15644565	Biotinidase deficiency
NM_000060.4(BTD):c.757C>T (p.Pro253Ser)	BTD	3	15644613	Biotinidase deficiency
NM_000060.4(BTD):c.896C>T (p.Ala299Val)	BTD	3	15644752	Biotinidase deficiency
NM_000060.4(BTD):c.929G>A (p.Gly310Glu)	BTD	3	15644785	Biotinidase deficiency
NM_000060.4(BTD):c.932G>A (p.Ser311Asn)	BTD	3	15644788	Biotinidase deficiency
NM_000060.4(BTD):c.934G>A (p.Gly312Ser)	BTD	3	15644790	Biotinidase deficiency
NM_000060.4(BTD):c.935G>A (p.Gly312Asp)	BTD	3	15644791	Biotinidase deficiency
NM_000060.4(BTD):c.1106C>T (p.Pro369Leu)	BTD	3	15644962	Biotinidase deficiency
NM_000060.4(BTD):c.1211C>T (p.Thr404Ile)	BTD	3	15645067	Biotinidase deficiency
NM_000060.4(BTD):c.1271G>A (p.Cys424Tyr)	BTD	3	15645127	Biotinidase deficiency
NM_000060.4(BTD):c.1333G>A (p.Gly445Arg)	BTD	3	15645189	Biotinidase deficiency
NM_000060.4(BTD):c.1339C>T (p.His447Tyr)	BTD	3	15645195	Biotinidase deficiency
NM_000060.4(BTD):c.1352G>A (p.Gly451Asp)	BTD	3	15645208	Biotinidase deficiency
NM_000060.4(BTD):c.1369G>A (p.Val457Met)	BTD	3	15645225	Biotinidase deficiency
NM_000060.4(BTD):c.1388G>A (p.Cys463Tyr)	BTD	3	15645244	Biotinidase deficiency
NM_000060.4(BTD):c.1463G>A (p.Gly488Asp)	BTD	3	15645319	Biotinidase deficiency
NM_000060.4(BTD):c.1610G>A (p.Gly537Glu)	BTD	3	15645466	Biotinidase deficiency
NM_000060.4(BTD):c.1613G>A (p.Arg538His)	BTD	3	15645469	Biotinidase deficiency
NM_000060.4(BTD):c.1432G>A (p.Ala478Thr)	BTD	3	15645288	Biotinidase deficiency
NM_000060.4(BTD):c.559C>T (p.Pro187Ser)	BTD	3	15644415	Biotinidase deficiency
NM_000060.4(BTD):c.734G>A (p.Cys245Tyr)	BTD	3	15644590	Biotinidase deficiency
NM_000060.4(BTD):c.184G>A (p.Val62Met)	BTD	3	15635563	Biotinidase deficiency
NM_000060.4(BTD):c.236G>A (p.Arg79His)	BTD	3	15635615	Biotinidase deficiency
NM_000060.4(BTD):c.1158G>A (p.Trp386Ter)	BTD	3	15645014	Biotinidase deficiency
NM_000060.4(BTD):c.758C>T (p.Pro253Leu)	BTD	3	15644614	Biotinidase deficiency
NM_000060.4(BTD):c.1237G>A (p.Gly413Ser)	BTD	3	15645093	Biotinidase deficiency
NM_000060.4(BTD):c.701C>T (p.Thr234Ile)	BTD	3	15644557	Biotinidase deficiency
NM_000060.4(BTD):c.1595C>T (p.Thr532Met)	BTD	3	15645451	Biotinidase deficiency;not provided
NM_000060.4(BTD):c.1612C>T (p.Arg538Cys)	BTD	3	15645468	Biotinidase deficiency;not provided
NM_000060.4(BTD):c.459G>A (p.Glu153Glu=)	BTD	3	15642057	Biotinidase deficiency;not provided
NM_000060.4(BTD):c.469C>T (p.Arg157Cys)	BTD	3	15644325	Biotinidase deficiency;not provided
NM_000060.4(BTD):c.511G>A (p.Ala171Thr)	BTD	3	15644367	Biotinidase deficiency;not provided
NM_000060.4(BTD):c.557G>A (p.Cys186Tyr)	BTD	3	15644413	Biotinidase deficiency;not provided
NM_000060.4(BTD):c.1489C>T (p.Pro497Ser)	BTD	3	15645345	Biotinidase deficiency;not provided
NM_000060.4(BTD):c.470G>A (p.Arg157His)	BTD	3	15644326	Biotinidase deficiency;not provided
NM_001282534.1(KCNK9):c.706G>A (p.Gly236Arg)	KCNK9	8	1396186777	Birk Barel mental retardation dysmorphism syndrome;not provided
NM_004328.4(BCS1L):c.550C>T (p.Arg184Cys)	BCS1L	2	218661848	Bjornstad syndrome with mild mitochondrial complex III deficiency;Mitochondrial complex III deficiency;not provided

NM_033360.3(KRAS):c.175G>A (p.Ala59Thr)	KRAS	12	25227349	Bladder cancer, transitional cell, somatic;Colorectal Neoplasms
NM_022162.2(NOD2):c.2798+158C>T	NOD2	16	50722863	Blau syndrome
NM_022162.2(NOD2):c.1405C>T (p.Leu469Phe)	NOD2	16	50711316	Blau syndrome;Sarcoidosis, early-onset
NM_022162.2(NOD2):c.1000C>T (p.Arg334Trp)	NOD2	16	50710911	Blau syndrome;Sarcoidosis, early-onset
NM_022162.2(NOD2):c.1147G>A (p.Glu383Lys)	NOD2	16	50711058	Blau syndrome;Sarcoidosis, early-onset
NM_022162.2(NOD2):c.1001G>A (p.Arg334Gln)	NOD2	16	50710912	Blau syndrome;Sarcoidosis, early-onset;not provided
NM_002017.4(FLI1):c.970C>T (p.Arg324Trp)	FLI1	11	128810599	Bleeding disorder platelet type macrothrombocytopenia
NM_001271010.1(FLI1):c.811C>T (p.Arg271Trp)	FLI1	11	128810638	BLEEDING DISORDER, PLATELET-TYPE, 21
NM_001271012.1(FLI1):c.431G>A (p.Arg144Gln)	FLI1	11	128810639	BLEEDING DISORDER, PLATELET-TYPE, 21
NM_023067.3(FOXL2):c.157C>T (p.Gln53Ter)	FOXL2	3	138946566	Blepharophimosis syndrome type 1
NM_023067.3(FOXL2):c.295C>T (p.Gln99Ter)	FOXL2	3	138946428	Blepharophimosis syndrome type 1
NM_023067.3(FOXL2):c.205G>A (p.Glu69Lys)	FOXL2	3	138946518	Blepharophimosis syndrome type 1
NM_023067.3(FOXL2):c.655C>T (p.Gln219Ter)	FOXL2	3	138946068	Blepharophimosis syndrome type 1;Blepharophimosis, ptosis, and epicanthus inversus
NM_023067.3(FOXL2):c.586C>T (p.Gln196Ter)	FOXL2	3	138946137	Blepharophimosis, ptosis, and epicanthus inversus
NM_023067.3(FOXL2):c.650C>T (p.Ser217Phe)	FOXL2	3	138946073	Blepharophimosis, ptosis, and epicanthus inversus
NM_023067.3(FOXL2):c.612G>A (p.Trp204Ter)	FOXL2	3	138946111	Blepharophimosis, ptosis, and epicanthus inversus
NM_023067.3(FOXL2):c.293G>A (p.Trp98Ter)	FOXL2	3	138946430	Blepharophimosis, ptosis, and epicanthus inversus
NM_023067.3(FOXL2):c.560G>A (p.Gly187Asp)	FOXL2	3	138946163	Blepharophimosis, ptosis, and epicanthus inversus;Premature ovarian failure 3
NM_025114.3(CEP290):c.2941C>T (p.Gln981Ter)	CEP290	12	88102888	Blindness;Central hypotonia;Molar tooth sign on MRI;Nystagmus
NM_022786.2(ARV1):c.565G>A (p.Gly189Arg)	ARV1	1	230995876	Blindness;Epileptic encephalopathy, early infantile, 38;Neurodegeneration
NM_025114.3(CEP290):c.4882C>T (p.Gln1628Ter)	CEP290	12	88083161	Blindness;Global developmental delay;Joubert syndrome 5
NM_000350.2(ABCA4):c.6445C>T (p.Arg2149Ter)	ABCA4	1	94000870	Blindness;Macular degeneration;Stargardt disease 1;Visual loss;not provided
NM_006563.4(KLF1):c.895C>T (p.His299Tyr)	KLF1	19	12885335	BLOOD GROUP--LUTHERAN INHIBITOR
NM_006563.4(KLF1):c.1012C>T (p.Pro338Ser)	KLF1	19	12884962	BLOOD GROUP--LUTHERAN INHIBITOR;Congenital dyserythropoietic anemia, type IV;Fetal hemoglobin quantitative trait locus 6
NM_000057.3(BLM):c.2643G>A (p.Trp881Ter)	BLM	15	90782909	Bloom syndrome
NM_000057.3(BLM):c.2887C>T (p.His963Tyr)	BLM	15	90790712	Bloom syndrome
NM_000057.3(BLM):c.3197G>A (p.Cys1066Tyr)	BLM	15	90794344	Bloom syndrome
NM_000057.3(BLM):c.3558+1G>A	BLM	15	90803721	Bloom syndrome
NM_000057.3(BLM):c.3847C>T (p.Gln1283Ter)	BLM	15	90809232	Bloom syndrome
NM_000057.3(BLM):c.3415C>T (p.Arg1139Ter)	BLM	15	90803577	Bloom syndrome
NM_000057.3(BLM):c.2875C>T (p.Arg959Ter)	BLM	15	90790700	Bloom syndrome
NM_000057.3(BLM):c.2098C>T (p.Gln700Ter)	BLM	15	90765319	Bloom syndrome;not provided
NM_000057.3(BLM):c.1642C>T (p.Gln548Ter)	BLM	15	90761015	Bloom syndrome;not provided
NM_000057.3(BLM):c.2695C>T (p.Arg899Ter)	BLM	15	90784953	Bloom syndrome;not provided
NM_181745.3(FFAR4):c.809G>A (p.Arg270His)	FFAR4	10	93587284	Body mass index quantitative trait locus 10
NM_003355.2(UCP2):c.-1245G>A	UCP2	11	73983709	Body mass index quantitative trait locus 4
NM_006947.3(SRP72):c.620G>A (p.Arg207His)	SRP72	4	56476680	Bone marrow failure syndrome 1
NM_020207.4(ERCC6L2):c.1963C>T (p.Arg655Ter)	ERCC6L2	9	95955996	Bone marrow failure syndrome 2
NM_001012339.2(DNAJC21):c.517C>T (p.Arg173Ter)	DNAJC21	5	34937404	Bone marrow failure syndrome 3;Inherited bone marrow failure syndrome
NM_005430.3(WNT1):c.703C>T (p.Arg235Trp)	WNT1	12	48981230	Bone mineral density quantitative trait locus 16
NM_001015877.1(PHF6):c.1024C>T (p.Arg342Ter)	PHF6	X	134425256	Borjeson-Forssman-Lehmann syndrome
NM_001015877.1(PHF6):c.134G>A (p.Cys45Tyr)	PHF6	X	134377751	Borjeson-Forssman-Lehmann syndrome
NM_001015877.1(PHF6):c.418G>A (p.Ala140Thr)	PHF6	X	134393952	Borjeson-Forssman-Lehmann syndrome
NM_001017995.2(SH3PXD2B):c.401+1G>A	SH3PXD2B	5	172382035	Borrone Di Rocco Crovato syndrome;Frank Ter Haar syndrome

NM_005654.5(NR2F1):c.335G>A (p.Arg112Lys)	NR2F1	5	93585358	Bosch-Boonstra-Schaaf optic atrophy syndrome
NM_005654.5(NR2F1):c.413G>A (p.Cys138Tyr)	NR2F1	5	93585436	Bosch-Boonstra-Schaaf optic atrophy syndrome
NM_000326.4(RLBP1):c.700C>T (p.Arg234Trp)	RLBP1	15	89210794	Bothnia retinal dystrophy;RLBP1-Related Disorders;Retinitis punctata albescens
NM_006702.4(PNPLA6):c.3029C>T (p.Thr1010Ile)	PNPLA6	19	7556502	Boucher Neuhauser syndrome
NM_006702.4(PNPLA6):c.3184G>A (p.Val1062Met)	PNPLA6	19	7557185	Boucher Neuhauser syndrome
NM_006702.4(PNPLA6):c.3295C>T (p.Arg1099Cys)	PNPLA6	19	7558861	Boucher Neuhauser syndrome
NM_001080463.1(DYNC2H1):c.10482+1G>A	DYNC2H1	11	103256241	Bowing of the long bones;Intrauterine growth retardation;Narrow chest
NM_001077488.3(GNAS):c.85C>T (p.Gln29Ter)	GNAS	20	58891811	Brachydactyly syndrome;Cognitive impairment;Hypocalcemia;Obesity;Round face;Short stature;Subcutaneous nodule;Tetany
NM_002181.3(IHH):c.283G>A (p.Glu95Lys)	IHH	2	219060185	Brachydactyly type A1
NM_002181.3(IHH):c.391G>A (p.Glu131Lys)	IHH	2	219057619	Brachydactyly type A1
NM_002181.3(IHH):c.298G>A (p.Asp100Asn)	IHH	2	219060170	Brachydactyly type A1
NM_002181.3(IHH):c.461C>T (p.Thr154Ile)	IHH	2	219057549	Brachydactyly type A1
NM_002181.3(IHH):c.383G>A (p.Arg128Gln)	IHH	2	219057627	Brachydactyly type A1
NM_001203.2(BMPR1B):c.1456C>T (p.Arg486Trp)	BMPR1B	4	95154620	Brachydactyly type A2
NM_001203.2(BMPR1B):c.1457G>A (p.Arg486Gln)	BMPR1B	4	95154621	Brachydactyly type A2
NM_000557.4(GDF5):c.1139G>A (p.Arg380Gln)	GDF5	20	35434276	Brachydactyly type A2
NM_004560.3(ROR2):c.2246G>A (p.Trp749Ter)	ROR2	9	91724248	Brachydactyly type B1
NM_004560.3(ROR2):c.2247G>A (p.Trp749Ter)	ROR2	9	91724247	Brachydactyly type B1
NM_005450.4(NOG):c.103C>T (p.Pro35Ser)	NOG	17	56594326	Brachydactyly type B2;Cushing's symphalangism;Stapes ankylosis with broad thumb and toes
NM_000557.4(GDF5):c.901C>T (p.Arg301Ter)	GDF5	20	35434514	Brachydactyly type C
NM_000921.4(PDE3A):c.1339G>A (p.Ala447Thr)	PDE3A	12	20616299	Brachydactyly with hypertension
NM_000921.4(PDE3A):c.1340C>T (p.Ala447Val)	PDE3A	12	20616300	Brachydactyly with hypertension
NM_000557.4(GDF5):c.1195C>T (p.Arg399Cys)	GDF5	20	35434220	Brachydactyly, type a1, c
NM_001203.2(BMPR1B):c.447-1G>A	BMPR1B	4	95124982	Brachydactyly, type a1, d
NM_021625.4(TRPV4):c.1858G>A (p.Val620Ile)	TRPV4	12	109792396	Brachyrachia (short spine dysplasia);Neuromuscular Diseases;Skeletal dysplasia
NM_021625.4(TRPV4):c.1847G>A (p.Arg616Gln)	TRPV4	12	109792407	Brachyrachia (short spine dysplasia);Skeletal dysplasia
NM_004722.3(AP4M1):c.952C>T (p.Arg318Ter)	AP4M1	7	100105981	Brain atrophy;CNS hypomyelination;Global developmental delay;Hypoplasia of the corpus callosum;Microcephaly;Spastic paraplegia 50, autosomal recessive
m.8393C>T	MT-ATP8	MT	8393	Brain pseudoatrophy, reversible, valproate-induced, susceptibility to
NM_001845.5(COL4A1):c.1685G>A (p.Gly562Glu)	COL4A1	13	110187181	Brain small vessel disease with hemorrhage
NM_001845.5(COL4A1):c.2159G>A (p.Gly720Asp)	COL4A1	13	110181326	Brain small vessel disease with hemorrhage
NM_001845.5(COL4A1):c.1769G>A (p.Gly590Glu)	COL4A1	13	110186513	Brain small vessel disease with hemorrhage
NM_001845.5(COL4A1):c.2194-1G>A	COL4A1	13	110179422	Brain small vessel disease with hemorrhage
NM_001845.5(COL4A1):c.2263G>A (p.Gly755Arg)	COL4A1	13	110179352	Brain small vessel disease with hemorrhage
NM_001845.5(COL4A1):c.2122G>A (p.Gly708Arg)	COL4A1	13	110181363	Brain small vessel disease with hemorrhage
NM_001845.5(COL4A1):c.2086G>A (p.Gly696Ser)	COL4A1	13	110183002	Brain small vessel disease with hemorrhage;not provided
NM_000038.5(APC):c.1621C>T (p.Gln541Ter)	APC	5	112828001	Brain tumor-polyposis syndrome 2;Familial adenomatous polyposis 1
NM_000038.5(APC):c.643C>T (p.Gln215Ter)	APC	5	112780901	Brain tumor-polyposis syndrome 2;Familial adenomatous polyposis 1;not provided
NM_001105.4(ACVR1):c.983G>A (p.Gly328Glu)	ACVR1	2	157766004	Brainstem glioma;Progressive myositis ossificans
NM_005881.3(BCKDK):c.466C>T (p.Arg156Ter)	BCKDK	16	31110247	Branched-chain keto acid dehydrogenase kinase deficiency
NM_001042425.1(TFAP2A):c.773G>A (p.Gly258Glu)	TFAP2A	6	10402590	Branchiooculofacial syndrome
NM_001042425.1(TFAP2A):c.874G>A (p.Glu292Lys)	TFAP2A	6	10400587	Branchiooculofacial syndrome
NM_001042425.1(TFAP2A):c.698G>A (p.Arg233Gln)	TFAP2A	6	10404562	Branchiooculofacial syndrome
NM_005982.3(SIX1):c.328C>T (p.Arg110Trp)	SIX1	14	60648862	Branchiootic syndrome 3

NM_000503.5(EYA1):c.1081C>T (p.Arg361Ter)	EYA1	8	71244662	Branchiootic syndrome;Melnick-Fraser syndrome
NM_175875.4(SIX5):c.472G>A (p.Ala158Thr)	SIX5	19	45768373	Branchiootorenal syndrome 2
NM_175875.4(SIX5):c.886G>A (p.Ala296Thr)	SIX5	19	45767073	Branchiootorenal syndrome 2
NM_175875.4(SIX5):c.1093G>A (p.Gly365Arg)	SIX5	19	45766866	Branchiootorenal syndrome 2
NM_007194.3(CHEK2):c.444+1G>A	CHEK2	22	28725242	Breast and colorectal cancer, susceptibility to;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5095C>T (p.Arg1699Trp)	BRCA1	17	43063931	Breast and colorectal cancer;Breast-ovarian cancer, familial 1;Familial cancer of breast;Fanconi anemia, complementation group A;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;Ovarian cancer;not provided
NM_000059.3(BRCA2):c.5206C>T (p.Gln1736Ter)	BRCA2	13	32339561	Breast cancer
NM_000059.3(BRCA2):c.2266C>T (p.Gln756Ter)	BRCA2	13	32336621	Breast cancer
NM_032043.2(BRIP1):c.897G>A (p.Met299Ile)	BRIP1	17	61808488	Breast cancer, early-onset
PHB:c.*729C>T	PHB	17	49404263	Breast cancer, susceptibility to
NM_000102.3(CYP17A1):c.715C>T (p.Arg239Ter)	CYP17A1	10	102834074	Breast cancer, susceptibility to;Combined partial 17-alpha-hydroxylase/17,20-lyase deficiency
NM_024675.3(PALB2):c.2323C>T (p.Gln775Ter)	PALB2	16	23629831	Breast cancer, susceptibility to;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_024675.3(PALB2):c.1027C>T (p.Gln343Ter)	PALB2	16	23635519	Breast cancer, susceptibility to;Familial cancer of breast;Hereditary cancer-predisposing syndrome;Pancreatic cancer 3
NM_024675.3(PALB2):c.2962C>T (p.Gln988Ter)	PALB2	16	23623003	Breast cancer, susceptibility to;Fanconi anemia, complementation group N;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.7558C>T (p.Arg2520Ter)	BRCA2	13	32356550	Breast cancer;Breast-ovarian cancer, familial 1;Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.213-1G>A	BRCA1	17	43104957	Breast cancer;Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.2572C>T (p.Gln858Ter)	BRCA1	17	43092959	Breast cancer;Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.3607C>T (p.Arg1203Ter)	BRCA1	17	43091924	Breast cancer;Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5503C>T (p.Arg1835Ter)	BRCA1	17	43045767	Breast cancer;Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.2059C>T (p.Gln687Ter)	BRCA1	17	43093472	Breast cancer;Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.4675+1G>A	BRCA1	17	43074330	Breast cancer;Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5251C>T (p.Arg1751Ter)	BRCA1	17	43057078	Breast cancer;Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5444G>A (p.Trp1815Ter)	BRCA1	17	43047666	Breast cancer;Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.9318G>A (p.Trp3106Ter)	BRCA2	13	32394750	Breast cancer;Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.9382C>T (p.Arg3128Ter)	BRCA2	13	32394814	Breast cancer;Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.274C>T (p.Gln92Ter)	BRCA2	13	32319283	Breast cancer;Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.6952C>T (p.Arg2318Ter)	BRCA2	13	32346841	Breast cancer;Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_007294.3(BRCA1):c.1687C>T (p.Gln563Ter)	BRCA1	17	43093844	Breast carcinoma;Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;Ovarian cancer;Ovarian neoplasm;not provided

NM_007294.3(BRCA1):c.5406+1G>A	BRCA1	17	43049120	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.5193+1G>A	BRCA1	17	43063332	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.2309C>T (p.Ser770Leu)	BRCA1	17	43093222	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.4986+1G>A	BRCA1	17	43070927	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.5194-1G>A	BRCA1	17	43057136	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.5407-1G>A	BRCA1	17	43047704	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.547+1G>A	BRCA1	17	43099774	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.628C>T (p.Gln210Ter)	BRCA1	17	43095888	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.2599C>T (p.Gln867Ter)	BRCA1	17	43092932	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.784C>T (p.Gln262Ter)	BRCA1	17	43094747	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.280C>T (p.Gln94Ter)	BRCA1	17	43104889	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.5542C>T (p.Gln1848Ter)	BRCA1	17	43045728	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.5161C>T (p.Gln1721Ter)	BRCA1	17	43063365	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.4573C>T (p.Gln1525Ter)	BRCA1	17	43074433	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.4270C>T (p.Gln1424Ter)	BRCA1	17	43082491	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.4225C>T (p.Gln1409Ter)	BRCA1	17	43082536	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.4066C>T (p.Gln1356Ter)	BRCA1	17	43091465	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.3679C>T (p.Gln1227Ter)	BRCA1	17	43091852	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.1918C>T (p.Gln640Ter)	BRCA1	17	43093613	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.963G>A (p.Trp321Ter)	BRCA1	17	43094568	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.718C>T (p.Gln240Ter)	BRCA1	17	43094813	Breast-ovarian cancer, familial 1
NM_007294.3(BRCA1):c.134+1G>A	BRCA1	17	43115725	Breast-ovarian cancer, familial 1
NM_000059.3(BRCA2):c.9196C>T (p.Gln3066Ter)	BRCA2	13	32380085	Breast-ovarian cancer, familial 1;Breast-ovarian cancer, familial 2;Fanconi anemia, complementation group D1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.8009C>T (p.Ser2670Leu)	BRCA2	13	32363211	Breast-ovarian cancer, familial 1;Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.9154C>T (p.Arg3052Trp)	BRCA2	13	32380043	Breast-ovarian cancer, familial 1;Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.1909+1G>A	BRCA2	13	32333388	Breast-ovarian cancer, familial 1;Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.212+1G>A	BRCA1	17	43106455	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.3991C>T (p.Gln1331Ter)	BRCA1	17	43091540	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.4097-1G>A	BRCA1	17	43091033	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.1059G>A (p.Trp353Ter)	BRCA1	17	43094472	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.1115G>A (p.Trp372Ter)	BRCA1	17	43094416	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.1138C>T (p.Gln380Ter)	BRCA1	17	43094393	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.140G>A (p.Cys47Tyr)	BRCA1	17	43106528	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.1612C>T (p.Gln538Ter)	BRCA1	17	43093919	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.1621C>T (p.Gln541Ter)	BRCA1	17	43093910	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.1630C>T (p.Gln544Ter)	BRCA1	17	43093901	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.178C>T (p.Gln60Ter)	BRCA1	17	43106490	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.1969C>T (p.Gln657Ter)	BRCA1	17	43093562	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.212G>A (p.Arg71Lys)	BRCA1	17	43106456	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.2275C>T (p.Gln759Ter)	BRCA1	17	43093256	Breast-ovarian cancer, familial 1;Familial cancer of breast

NM_007294.3(BRCA1):c.2410C>T (p.Gln804Ter)	BRCA1	17	43093121	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.2869C>T (p.Gln957Ter)	BRCA1	17	43092662	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.2923C>T (p.Gln975Ter)	BRCA1	17	43092608	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.302-1G>A	BRCA1	17	43104262	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.3268C>T (p.Gln1090Ter)	BRCA1	17	43092263	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.3430C>T (p.Gln1144Ter)	BRCA1	17	43092101	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.3544C>T (p.Gln1182Ter)	BRCA1	17	43091987	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.4075C>T (p.Gln1359Ter)	BRCA1	17	43091456	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.4185G>A (p.Gln1395=)	BRCA1	17	43090944	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.4186C>T (p.Gln1396Ter)	BRCA1	17	43082575	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.4201C>T (p.Gln1401Ter)	BRCA1	17	43082560	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.4399C>T (p.Gln1467Ter)	BRCA1	17	43076573	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.441+1G>A	BRCA1	17	43104121	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.4552C>T (p.Gln1518Ter)	BRCA1	17	43074454	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5054C>T (p.Thr1685Ile)	BRCA1	17	43067628	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.514C>T (p.Gln172Ter)	BRCA1	17	43099808	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5153-1G>A	BRCA1	17	43063374	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5239C>T (p.Gln1747Ter)	BRCA1	17	43057090	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5266C>T (p.Gln1756Ter)	BRCA1	17	43057063	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5335C>T (p.Gln1779Ter)	BRCA1	17	43049192	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5345G>A (p.Trp1782Ter)	BRCA1	17	43049182	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5467G>A (p.Ala1823Thr)	BRCA1	17	43047643	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5511G>A (p.Trp1837Ter)	BRCA1	17	43045759	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.5536C>T (p.Gln1846Ter)	BRCA1	17	43045734	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.55C>T (p.Gln19Ter)	BRCA1	17	43124042	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.671-1G>A	BRCA1	17	43094861	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.80+1G>A	BRCA1	17	43124016	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.949C>T (p.Gln317Ter)	BRCA1	17	43094582	Breast-ovarian cancer, familial 1;Familial cancer of breast
NM_007294.3(BRCA1):c.928C>T (p.Gln310Ter)	BRCA1	17	43094603	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.5117G>A (p.Gly1706Glu)	BRCA1	17	43063909	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5136G>A (p.Trp1712Ter)	BRCA1	17	43063890	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.4327C>T (p.Arg1443Ter)	BRCA1	17	43082434	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided;not specified
NM_007294.3(BRCA1):c.1471C>T (p.Gln491Ter)	BRCA1	17	43094060	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.1576C>T (p.Gln526Ter)	BRCA1	17	43093955	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.160C>T (p.Gln54Ter)	BRCA1	17	43106508	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.2683C>T (p.Gln895Ter)	BRCA1	17	43092848	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.2761C>T (p.Gln921Ter)	BRCA1	17	43092770	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.3895C>T (p.Gln1299Ter)	BRCA1	17	43091636	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.4339C>T (p.Gln1447Ter)	BRCA1	17	43082422	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.4372C>T (p.Gln1458Ter)	BRCA1	17	43076600	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.5153G>A (p.Trp1718Ter)	BRCA1	17	43063373	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome

NM_007294.3(BRCA1):c.5445G>A (p.Trp1815Ter)	BRCA1	17	43047665	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.5510G>A (p.Trp1837Ter)	BRCA1	17	43045760	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.4675G>A (p.Glu1559Lys)	BRCA1	17	43074331	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5074G>A (p.Asp1692Asn)	BRCA1	17	43067608	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5346G>A (p.Trp1782Ter)	BRCA1	17	43049181	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.1116G>A (p.Trp372Ter)	BRCA1	17	43094415	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.1999C>T (p.Gln667Ter)	BRCA1	17	43093532	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.4183C>T (p.Gln1395Ter)	BRCA1	17	43090946	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.4810C>T (p.Gln1604Ter)	BRCA1	17	43071104	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.850C>T (p.Gln284Ter)	BRCA1	17	43094681	Breast-ovarian cancer, familial 1;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.1058G>A (p.Trp353Ter)	BRCA1	17	43094473	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.131G>A (p.Cys44Tyr)	BRCA1	17	43115729	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.1600C>T (p.Gln534Ter)	BRCA1	17	43093931	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.3286C>T (p.Gln1096Ter)	BRCA1	17	43092245	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.3403C>T (p.Gln1135Ter)	BRCA1	17	43092128	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.34C>T (p.Gln12Ter)	BRCA1	17	43124063	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.4258C>T (p.Gln1420Ter)	BRCA1	17	43082503	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.4485-1G>A	BRCA1	17	43074522	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.4609C>T (p.Gln1537Ter)	BRCA1	17	43074397	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.5075-1G>A	BRCA1	17	43063952	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.5154G>A (p.Trp1718Ter)	BRCA1	17	43063372	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided
NM_007294.3(BRCA1):c.5431C>T (p.Gln1811Ter)	BRCA1	17	43047679	Breast-ovarian cancer, familial 1;Familial cancer of breast;not provided;not specified
NM_007294.3(BRCA1):c.4484+1G>A	BRCA1	17	43076487	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.241C>T (p.Gln81Ter)	BRCA1	17	43104928	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.3331C>T (p.Gln1111Ter)	BRCA1	17	43092200	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.3967C>T (p.Gln1323Ter)	BRCA1	17	43091564	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.415C>T (p.Gln139Ter)	BRCA1	17	43104148	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.505C>T (p.Gln169Ter)	BRCA1	17	43099817	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.5194-12G>A	BRCA1	17	43057147	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.5212G>A (p.Gly1738Arg)	BRCA1	17	43057117	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.5278-1G>A	BRCA1	17	43051118	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.5332+1G>A	BRCA1	17	43051062	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.4676-1G>A	BRCA1	17	43071239	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.5468-1G>A	BRCA1	17	43045803	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome
NM_007294.3(BRCA1):c.1480C>T (p.Gln494Ter)	BRCA1	17	43094051	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome

NM_007294.3(BRCA1):c.2563C>T (p.Gln855Ter)	BRCA1	17	43092968	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.1066C>T (p.Gln356Ter)	BRCA1	17	43094465	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.3718C>T (p.Gln1240Ter)	BRCA1	17	43091813	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.3817C>T (p.Gln1273Ter)	BRCA1	17	43091714	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.3937C>T (p.Gln1313Ter)	BRCA1	17	43091594	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.4096+1G>A	BRCA1	17	43091434	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.4357+1G>A	BRCA1	17	43082403	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5074+1G>A	BRCA1	17	43067607	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5277+1G>A	BRCA1	17	43057051	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5467+1G>A	BRCA1	17	43047642	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.191G>A (p.Cys64Tyr)	BRCA1	17	43106477	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.2338C>T (p.Gln780Ter)	BRCA1	17	43093193	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.3598C>T (p.Gln1200Ter)	BRCA1	17	43091933	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.3841C>T (p.Gln1281Ter)	BRCA1	17	43091690	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.4222C>T (p.Gln1408Ter)	BRCA1	17	43082539	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.4524G>A (p.Trp1508Ter)	BRCA1	17	43074482	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.5353C>T (p.Gln1785Ter)	BRCA1	17	43049174	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.81-1G>A	BRCA1	17	43115780	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_007294.3(BRCA1):c.962G>A (p.Trp321Ter)	BRCA1	17	43094569	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided;not specified
NM_007294.3(BRCA1):c.220C>T (p.Gln74Ter)	BRCA1	17	43104949	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;not provided
NM_007294.3(BRCA1):c.2713C>T (p.Gln905Ter)	BRCA1	17	43092818	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;not provided
NM_007294.3(BRCA1):c.2800C>T (p.Gln934Ter)	BRCA1	17	43092731	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;not provided
NM_007294.3(BRCA1):c.4612C>T (p.Gln1538Ter)	BRCA1	17	43074394	Breast-ovarian cancer, familial 1;Hereditary breast and ovarian cancer syndrome;not provided
NM_007294.3(BRCA1):c.3352C>T (p.Gln1118Ter)	BRCA1	17	43092179	Breast-ovarian cancer, familial 1;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.5152+1G>A	BRCA1	17	43063873	Breast-ovarian cancer, familial 1;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.301+1G>A	BRCA1	17	43104867	Breast-ovarian cancer, familial 1;Hereditary cancer-predisposing syndrome

NM_007294.3(BRCA1):c.4834C>T (p.Gln1612Ter)	BRCA1	17	43071080	Breast-ovarian cancer, familial 1;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.4523G>A (p.Trp1508Ter)	BRCA1	17	43074483	Breast-ovarian cancer, familial 1;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.5135G>A (p.Trp1712Ter)	BRCA1	17	43063891	Breast-ovarian cancer, familial 1;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.1155G>A (p.Trp385Ter)	BRCA1	17	43094376	Breast-ovarian cancer, familial 1;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.442C>T (p.Gln148Ter)	BRCA1	17	43099880	Breast-ovarian cancer, familial 1;Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.4987-1G>A	BRCA1	17	43067696	Breast-ovarian cancer, familial 1;not provided
NM_000059.3(BRCA2):c.316+1G>A	BRCA2	13	32319326	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.7235C>T (p.Thr2412Ile)	BRCA2	13	32355088	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.9573G>A (p.Trp3191Ter)	BRCA2	13	32396969	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.8633-1G>A	BRCA2	13	32376669	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.1945C>T (p.Gln649Ter)	BRCA2	13	32336300	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.217C>T (p.Gln73Ter)	BRCA2	13	32319226	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.425G>A (p.Ser142Asn)	BRCA2	13	32325184	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.523C>T (p.Gln175Ter)	BRCA2	13	32326505	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.2548C>T (p.Gln850Ter)	BRCA2	13	32336903	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.2905C>T (p.Gln969Ter)	BRCA2	13	32337260	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.4689G>A (p.Trp1563Ter)	BRCA2	13	32339044	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.4972C>T (p.Gln1658Ter)	BRCA2	13	32339327	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.11184G>A (p.Trp3595Ter)	BRCA2	13	32332662	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.2137C>T (p.Gln713Ter)	BRCA2	13	32336492	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.3217C>T (p.Gln1073Ter)	BRCA2	13	32337572	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.3523C>T (p.Gln1175Ter)	BRCA2	13	32337878	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.4783C>T (p.Gln1595Ter)	BRCA2	13	32339138	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.5800C>T (p.Gln1934Ter)	BRCA2	13	32340155	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.6478C>T (p.Gln2160Ter)	BRCA2	13	32340833	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.7033C>T (p.Gln2345Ter)	BRCA2	13	32354886	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.7495C>T (p.Gln2499Ter)	BRCA2	13	32356487	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.7501C>T (p.Gln2501Ter)	BRCA2	13	32356493	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.7887G>A (p.Trp2629Ter)	BRCA2	13	32362604	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.8910G>A (p.Trp2970Ter)	BRCA2	13	32379472	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.9139C>T (p.Gln3047Ter)	BRCA2	13	32380028	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.9739C>T (p.Gln3247Ter)	BRCA2	13	32398252	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.6937+1G>A	BRCA2	13	32344654	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.6938-1G>A	BRCA2	13	32346826	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.7977-1G>A	BRCA2	13	32363178	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.8332-1G>A	BRCA2	13	32370401	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.9118-1G>A	BRCA2	13	32380006	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.-39-1G>A	BRCA2	13	32316421	Breast-ovarian cancer, familial 2
NM_000059.3(BRCA2):c.476-1G>A	BRCA2	13	32326241	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.582G>A (p.Trp194Ter)	BRCA2	13	32326564	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.7963C>T (p.Gln2655Ter)	BRCA2	13	32362680	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8695C>T (p.Gln2899Ter)	BRCA2	13	32376732	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8869C>T (p.Gln2957Ter)	BRCA2	13	32379431	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.1117C>T (p.Gln373Ter)	BRCA2	13	32332595	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.1825C>T (p.Gln609Ter)	BRCA2	13	32333303	Breast-ovarian cancer, familial 2;Familial cancer of breast

NM_000059.3(BRCA2):c.2455C>T (p.Gln819Ter)	BRCA2	13	32336810	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.2881C>T (p.Gln961Ter)	BRCA2	13	32337236	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.3265C>T (p.Gln1089Ter)	BRCA2	13	32337620	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.3283C>T (p.Gln1095Ter)	BRCA2	13	32337638	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.3442C>T (p.Gln1148Ter)	BRCA2	13	32337797	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.3871C>T (p.Gln1291Ter)	BRCA2	13	32338226	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.439C>T (p.Gln147Ter)	BRCA2	13	32326114	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.4525C>T (p.Gln1509Ter)	BRCA2	13	32338880	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.475+1G>A	BRCA2	13	32326151	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.5344C>T (p.Gln1782Ter)	BRCA2	13	32339699	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.5404C>T (p.Gln1802Ter)	BRCA2	13	32339759	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.5773C>T (p.Gln1925Ter)	BRCA2	13	32340128	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.5992C>T (p.Gln1998Ter)	BRCA2	13	32340347	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.632-1G>A	BRCA2	13	32329442	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.6469C>T (p.Gln2157Ter)	BRCA2	13	32340824	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.7261C>T (p.Gln2421Ter)	BRCA2	13	32355114	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.7303C>T (p.Gln2435Ter)	BRCA2	13	32355156	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.7471C>T (p.Gln2491Ter)	BRCA2	13	32356463	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.7681C>T (p.Gln2561Ter)	BRCA2	13	32357805	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.7738C>T (p.Gln2580Ter)	BRCA2	13	32357862	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.7886G>A (p.Trp2629Ter)	BRCA2	13	32362603	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8140C>T (p.Gln2714Ter)	BRCA2	13	32363342	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8363G>A (p.Trp2788Ter)	BRCA2	13	32370433	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8485C>T (p.Gln2829Ter)	BRCA2	13	32370555	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8572C>T (p.Gln2858Ter)	BRCA2	13	32371040	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8632+1G>A	BRCA2	13	32371101	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8754+1G>A	BRCA2	13	32376792	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8773C>T (p.Gln2925Ter)	BRCA2	13	32379335	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8821C>T (p.Gln2941Ter)	BRCA2	13	32379383	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.9109C>T (p.Gln3037Ter)	BRCA2	13	32379905	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.9117+1G>A	BRCA2	13	32379914	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.9256+1G>A	BRCA2	13	32380146	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.9317G>A (p.Trp3106Ter)	BRCA2	13	32394749	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.9466C>T (p.Gln3156Ter)	BRCA2	13	32394898	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.9501+1G>A	BRCA2	13	32394934	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.9501G>A (p.Glu3167=)	BRCA2	13	32394933	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.9572G>A (p.Trp3191Ter)	BRCA2	13	32396968	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.8490G>A (p.Trp2830Ter)	BRCA2	13	32370958	Breast-ovarian cancer, familial 2;Familial cancer of breast
NM_000059.3(BRCA2):c.5980C>T (p.Gln1994Ter)	BRCA2	13	32340335	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.7721G>A (p.Trp2574Ter)	BRCA2	13	32357845	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.196C>T (p.Gln66Ter)	BRCA2	13	32319205	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome

NM_000059.3(BRCA2):c.7618-1G>A	BRCA2	13	32357741	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.8489G>A (p.Trp2830Ter)	BRCA2	13	32370957	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.7857G>A (p.Trp2619Ter)	BRCA2	13	32362574	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.1261C>T (p.Gln421Ter)	BRCA2	13	32332739	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.1456C>T (p.Gln486Ter)	BRCA2	13	32332934	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.3319C>T (p.Gln1107Ter)	BRCA2	13	32337674	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.517-1G>A	BRCA2	13	32326498	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.5791C>T (p.Gln1931Ter)	BRCA2	13	32340146	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.6070C>T (p.Gln2024Ter)	BRCA2	13	32340425	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.7024C>T (p.Gln2342Ter)	BRCA2	13	32354877	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.8754G>A (p.Glu2918=)	BRCA2	13	32376791	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.961C>T (p.Gln321Ter)	BRCA2	13	32332439	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.8331+1G>A	BRCA2	13	32363534	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.9380G>A (p.Trp3127Ter)	BRCA2	13	32394812	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.475G>A (p.Val159Met)	BRCA2	13	32326150	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.631G>A (p.Val211Ile)	BRCA2	13	32326613	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.7976+1G>A	BRCA2	13	32362694	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.8364G>A (p.Trp2788Ter)	BRCA2	13	32370434	Breast-ovarian cancer, familial 2;Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.7758G>A (p.Trp2586Ter)	BRCA2	13	32357882	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.2224C>T (p.Gln742Ter)	BRCA2	13	32336579	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.5101C>T (p.Gln1701Ter)	BRCA2	13	32339456	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.516+1G>A	BRCA2	13	32326283	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.5959C>T (p.Gln1987Ter)	BRCA2	13	32340314	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.67+1G>A	BRCA2	13	32316528	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.7060C>T (p.Gln2354Ter)	BRCA2	13	32354913	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.8953+1G>A	BRCA2	13	32379516	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.9100C>T (p.Gln3034Ter)	BRCA2	13	32379896	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.9148C>T (p.Gln3050Ter)	BRCA2	13	32380037	Breast-ovarian cancer, familial 2;Familial cancer of breast;not provided
NM_000059.3(BRCA2):c.9883C>T (p.Gln3295Ter)	BRCA2	13	32398396	Breast-ovarian cancer, familial 2;Familial cancer of breast;not specified
NM_000059.3(BRCA2):c.631+1G>A	BRCA2	13	32326614	Breast-ovarian cancer, familial 2;Fanconi anemia, complementation group D1;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.1414C>T (p.Gln472Ter)	BRCA2	13	32332892	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.1689G>A (p.Trp563Ter)	BRCA2	13	32333167	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.581G>A (p.Trp194Ter)	BRCA2	13	32326563	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.6490C>T (p.Gln2164Ter)	BRCA2	13	32340845	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.7805+1G>A	BRCA2	13	32357930	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.7856G>A (p.Trp2619Ter)	BRCA2	13	32362573	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome

NM_000059.3(BRCA2):c.8970G>A (p.Trp2990Ter)	BRCA2	13	32379766	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.92G>A (p.Trp31Ter)	BRCA2	13	32319101	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.9376C>T (p.Gln3126Ter)	BRCA2	13	32394808	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.93G>A (p.Trp31Ter)	BRCA2	13	32319102	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.7008-1G>A	BRCA2	13	32354860	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.1189C>T (p.Gln397Ter)	BRCA2	13	32332667	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.2818C>T (p.Gln940Ter)	BRCA2	13	32337173	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.2979G>A (p.Trp993Ter)	BRCA2	13	32337334	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.3166C>T (p.Gln1056Ter)	BRCA2	13	32337521	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.3G>A (p.Met1Ile)	BRCA2	13	32316463	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.4285C>T (p.Gln1429Ter)	BRCA2	13	32338640	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.6025C>T (p.Gln2009Ter)	BRCA2	13	32340380	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.772C>T (p.Gln258Ter)	BRCA2	13	32331009	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.7877G>A (p.Trp2626Ter)	BRCA2	13	32362594	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.3109C>T (p.Gln1037Ter)	BRCA2	13	32337464	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.4222C>T (p.Gln1408Ter)	BRCA2	13	32338577	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.7480C>T (p.Arg2494Ter)	BRCA2	13	32356472	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.7878G>A (p.Trp2626Ter)	BRCA2	13	32362595	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.8488-1G>A	BRCA2	13	32370955	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.9076C>T (p.Gln3026Ter)	BRCA2	13	32379872	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.9117G>A (p.Pro3039=)	BRCA2	13	32379913	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.1855C>T (p.Gln619Ter)	BRCA2	13	32333333	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.4111C>T (p.Gln1371Ter)	BRCA2	13	32338466	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.5656C>T (p.Gln1886Ter)	BRCA2	13	32340011	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.7757G>A (p.Trp2586Ter)	BRCA2	13	32357881	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided

NM_000059.3(BRCA2):c.8243G>A (p.Gly2748Asp)	BRCA2	13	32363445	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.8878C>T (p.Gln2960Ter)	BRCA2	13	32379440	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.8487+1G>A	BRCA2	13	32370558	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;Hereditary cancer-predisposing syndrome;not provided;not specified
NM_000059.3(BRCA2):c.8677C>T (p.Gln2893Ter)	BRCA2	13	32376714	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.250C>T (p.Gln84Ter)	BRCA2	13	32319259	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.6124C>T (p.Gln2042Ter)	BRCA2	13	32340479	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.7617+1G>A	BRCA2	13	32356610	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.8575C>T (p.Gln2859Ter)	BRCA2	13	32371043	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.8174G>A (p.Trp2725Ter)	BRCA2	13	32363376	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.3187C>T (p.Gln1063Ter)	BRCA2	13	32337542	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.9381G>A (p.Trp3127Ter)	BRCA2	13	32394813	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.2095C>T (p.Gln699Ter)	BRCA2	13	32336450	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not provided
NM_000059.3(BRCA2):c.793+1G>A	BRCA2	13	32331031	Breast-ovarian cancer, familial 2;Hereditary breast and ovarian cancer syndrome;not specified
NM_000059.3(BRCA2):c.1642C>T (p.Gln548Ter)	BRCA2	13	32333120	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.8608C>T (p.Gln2870Ter)	BRCA2	13	32371076	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.425+1G>A	BRCA2	13	32325185	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.3412C>T (p.Gln1138Ter)	BRCA2	13	32337767	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.4246C>T (p.Gln1416Ter)	BRCA2	13	32338601	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.6475C>T (p.Gln2159Ter)	BRCA2	13	32340830	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.7366C>T (p.Gln2456Ter)	BRCA2	13	32355219	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.7516C>T (p.Gln2506Ter)	BRCA2	13	32356508	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome
NM_000059.3(BRCA2):c.8969G>A (p.Trp2990Ter)	BRCA2	13	32379765	Breast-ovarian cancer, familial 2;Hereditary cancer-predisposing syndrome;not provided
NM_000059.3(BRCA2):c.6487C>T (p.Gln2163Ter)	BRCA2	13	32340842	Breast-ovarian cancer, familial 2;not provided
NM_000059.3(BRCA2):c.2978G>A (p.Trp993Ter)	BRCA2	13	32337333	Breast-ovarian cancer, familial 2;not provided
NM_000059.3(BRCA2):c.7615C>T (p.Gln2539Ter)	BRCA2	13	32356607	Breast-ovarian cancer, familial 2;not provided
NM_000059.3(BRCA2):c.9106C>T (p.Gln3036Ter)	BRCA2	13	32379902	Breast-ovarian cancer, familial 2;not provided
NM_058216.2(RAD51C):c.397C>T (p.Gln133Ter)	RAD51C	17	58695182	Breast-ovarian cancer, familial 3;Fanconi anemia, complementation group O;Hereditary cancer-predisposing syndrome;not provided
NM_058216.2(RAD51C):c.97C>T (p.Gln33Ter)	RAD51C	17	58692740	Breast-ovarian cancer, familial 3;Fanconi anemia, complementation group O;Hereditary cancer-predisposing syndrome;not provided
NM_058216.2(RAD51C):c.837+1G>A	RAD51C	17	58709991	Breast-ovarian cancer, familial 3;Hereditary cancer-predisposing syndrome
NM_002878.3(RAD51D):c.576+1G>A	RAD51D	17	35106385	Breast-ovarian cancer, familial 4
NM_002878.3(RAD51D):c.803G>A (p.Trp268Ter)	RAD51D	17	35101301	Breast-ovarian cancer, familial 4
NM_002878.3(RAD51D):c.547C>T (p.Gln183Ter)	RAD51D	17	35106415	Breast-ovarian cancer, familial 4;Hereditary cancer-predisposing syndrome
NM_002878.3(RAD51D):c.556C>T (p.Arg186Ter)	RAD51D	17	35106406	Breast-ovarian cancer, familial 4;Hereditary cancer-predisposing syndrome;not provided
NM_002878.3(RAD51D):c.757C>T (p.Arg253Ter)	RAD51D	17	35101347	Breast-ovarian cancer, familial 4;Hereditary cancer-predisposing syndrome;not provided
NM_002878.3(RAD51D):c.694C>T (p.Arg232Ter)	RAD51D	17	35103298	Breast-ovarian cancer, familial 4;Hereditary cancer-predisposing syndrome;not provided
NM_002878.3(RAD51D):c.451C>T (p.Gln151Ter)	RAD51D	17	35107017	Breast-ovarian cancer, familial 4;Hereditary cancer-predisposing syndrome;not provided
NM_018699.3(PRDM5):c.1768C>T (p.Arg590Ter)	PRDM5	4	120695236	Brittle cornea syndrome 2
NM_001005463.2(EBF3):c.512G>A (p.Gly171Asp)	EBF3	10	129957300	Broad-based gait;Developmental regression;Generalized hypotonia;Neurogenic bladder;Renal dysplasia
NM_004320.4(ATP2A1):c.592C>T (p.Arg198Ter)	ATP2A1	16	28887236	Brody myopathy

NM_004320.4(ATP2A1):c.2366C>T (p.Pro789Leu)	ATP2A1	16	28902228	Brody myopathy
NM_000336.2(SCNN1B):c.800C>T (p.Pro267Leu)	SCNN1B	16	23367879	Bronchiectasis with or without elevated sweat chloride 1
NM_000336.2(SCNN1B):c.1615G>A (p.Glu539Lys)	SCNN1B	16	23380493	Bronchiectasis with or without elevated sweat chloride 1
NM_001038.5(SCNN1A):c.340G>A (p.Val114Ile)	SCNN1A	12	6374444	Bronchiectasis with or without elevated sweat chloride 2
NM_001038.5(SCNN1A):c.241C>T (p.Arg81Cys)	SCNN1A	12	6374543	Bronchiectasis with or without elevated sweat chloride 2
NM_033409.3(SLC52A3):c.394C>T (p.Arg132Trp)	SLC52A3	20	765381	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.106G>A (p.Glu36Lys)	SLC52A3	20	765569	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.1255G>A (p.Val419Met)	SLC52A3	20	761181	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.955C>T (p.Pro319Ser)	SLC52A3	20	763616	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.935C>T (p.Ala312Val)	SLC52A3	20	763636	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.634C>T (p.Arg212Cys)	SLC52A3	20	763937	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.211G>A (p.Glu71Lys)	SLC52A3	20	765564	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.160G>A (p.Gly54Arg)	SLC52A3	20	765615	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.1293G>A (p.Trp431Ter)	SLC52A3	20	761143	Brown-Vialetto-Van Laere syndrome 1
NM_033409.3(SLC52A3):c.796C>T (p.Arg266Trp)	SLC52A3	20	763775	Brown-Vialetto-Van Laere syndrome 1;not provided
NM_024531.4(SLC52A2):c.1255G>A (p.Gly419Ser)	SLC52A2	8	144360932	Brown-Vialetto-Van Laere syndrome 2
NM_024531.4(SLC52A2):c.383C>T (p.Ser128Leu)	SLC52A2	8	144359875	Brown-Vialetto-Van Laere syndrome 2
NM_024531.4(SLC52A2):c.700C>T (p.Gln234Ter)	SLC52A2	8	144360192	Brown-Vialetto-Van Laere syndrome 2
NM_024531.4(SLC52A2):c.865C>T (p.Leu289=)	SLC52A2	8	144360357	Brown-Vialetto-Van Laere syndrome 2
NM_024531.4(SLC52A2):c.1088C>T (p.Pro363Leu)	SLC52A2	8	144360676	Brown-Vialetto-Van Laere syndrome 2
NM_024531.4(SLC52A2):c.916G>A (p.Gly306Arg)	SLC52A2	8	144360408	Brown-Vialetto-Van Laere syndrome 2;not provided
NM_024531.4(SLC52A2):c.155C>T (p.Ser52Phe)	SLC52A2	8	144359647	Brown-Vialetto-Van Laere syndrome 2;not provided
NM_001253815.1(SLC52A2):c.808C>T (p.Gln270Ter)	SLC52A2	8	144360300	Brown-Vialetto-Van Laere syndrome 2;not provided
NM_024531.4(SLC52A2):c.1258G>A (p.Ala420Thr)	SLC52A2	8	144360935	Brown-Vialetto-Van Laere syndrome 2;not provided
NM_021939.3(FKBP10):c.344G>A (p.Arg115Gln)	FKBP10	17	41817156	Bruck syndrome 1
NM_021939.3(FKBP10):c.337G>A (p.Glu113Lys)	FKBP10	17	41817149	Bruck syndrome 1
NM_000935.2(PLOD2):c.1823C>T (p.Thr608Ile)	PLOD2	3	146071386	Bruck syndrome 2
NM_000935.2(PLOD2):c.1793G>A (p.Arg598His)	PLOD2	3	146071416	Bruck syndrome 2
NM_198056.2(SCN5A):c.362G>A (p.Arg121Gln)	SCN5A	3	38630341	Brugada syndrome
NM_198056.2(SCN5A):c.481G>A (p.Glu161Lys)	SCN5A	3	38622401	Brugada syndrome
NM_198056.2(SCN5A):c.4772G>A (p.Trp1591Ter)	SCN5A	3	38554320	Brugada syndrome
NM_198056.2(SCN5A):c.2636G>A (p.Trp879Ter)	SCN5A	3	38585842	Brugada syndrome
NM_198056.2(SCN5A):c.2933G>A (p.Trp978Ter)	SCN5A	3	38581226	Brugada syndrome
NM_198056.2(SCN5A):c.3285G>A (p.Trp1095Ter)	SCN5A	3	38579439	Brugada syndrome
NM_000335.4(SCN5A):c.4259G>A (p.Trp1420Ter)	SCN5A	3	38557268	Brugada syndrome 1
NM_000335.4(SCN5A):c.5126C>T (p.Ser1709Leu)	SCN5A	3	38551243	Brugada syndrome 1;Cardiovascular phenotype;Paroxysmal familial ventricular fibrillation 1;Ventricular fibrillation;not provided
NM_015141.3(GPD1L):c.839C>T (p.Ala280Val)	GPD1L	3	32159096	Brugada syndrome 2;Cardiovascular phenotype;Death in infancy;not provided
NM_201590.2(CACNB2):c.1442C>T (p.Ser481Leu)	CACNB2	10	18539345	Brugada syndrome 4
NM_201590.2(CACNB2):c.32C>T (p.Thr11Ile)	CACNB2	10	18340958	Brugada syndrome 4
NM_001037.4(SCN1B):c.448+88G>A	SCN1B	19	35033827	Brugada syndrome 5;not provided
NM_004980.4(KCND3):c.1798G>A (p.Gly600Arg)	KCND3	1	111776247	Brugada syndrome 9
NM_004980.4(KCND3):c.1348C>T (p.Leu450Phe)	KCND3	1	111780713	Brugada syndrome 9;not provided
NM_198056.2(SCN5A):c.1100G>A (p.Arg367His)	SCN5A	3	38606709	Brugada syndrome;Brugada syndrome 1
NM_198056.2(SCN5A):c.2204C>T (p.Ala735Val)	SCN5A	3	38597787	Brugada syndrome;Brugada syndrome 1

NM_000335.4(SCN5A):c.3781G>A (p.Gly1261Ser)	SCN5A	3	38566465	Brugada syndrome;Brugada syndrome 1
NM_198056.2(SCN5A):c.4859C>T (p.Thr1620Met)	SCN5A	3	38551513	Brugada syndrome;Brugada syndrome 1
NM_198056.2(SCN5A):c.845G>A (p.Arg282His)	SCN5A	3	38609823	Brugada syndrome;Brugada syndrome 1
NM_198056.2(SCN5A):c.4222G>A (p.Gly1408Arg)	SCN5A	3	38560170	Brugada syndrome;Brugada syndrome 1;Cardiac conduction defect, nonspecific;Sick sinus syndrome 1, autosomal recessive;not provided
NM_198056.2(SCN5A):c.5350G>A (p.Glu1784Lys)	SCN5A	3	38551022	Brugada syndrome;Brugada syndrome 1;Cardiovascular phenotype;Congenital long QT syndrome;Long QT syndrome 3;Sinus node disease;not provided
NM_198056.2(SCN5A):c.5770G>A (p.Ala1924Thr)	SCN5A	3	38550602	Brugada syndrome;Brugada syndrome 1;not provided
NM_000719.6(CACNA1C):c.116C>T (p.Ala39Val)	CACNA1C	12	2115290	Brugada syndrome;Brugada syndrome 3
NM_198056.2(SCN5A):c.4931G>A (p.Arg1644His)	SCN5A	3	38551441	Brugada syndrome;Cardiovascular phenotype;Congenital long QT syndrome;Long QT syndrome 3;not provided
NM_198056.2(SCN5A):c.2254G>A (p.Gly752Arg)	SCN5A	3	38597737	Brugada syndrome;Cardiovascular phenotype;not provided
NM_198056.2(SCN5A):c.665G>A (p.Arg222Gln)	SCN5A	3	38613781	Brugada syndrome;Congenital long QT syndrome;Dilated cardiomyopathy 1E;Primary dilated cardiomyopathy;not provided
NM_000335.4(SCN5A):c.1231G>A (p.Val411Met)	SCN5A	3	38606058	Brugada syndrome;Congenital long QT syndrome;Long QT syndrome;Long QT syndrome 3;not provided
NM_198056.2(SCN5A):c.2440C>T (p.Arg814Trp)	SCN5A	3	38586038	Brugada syndrome;Dilated cardiomyopathy;Primary dilated cardiomyopathy;not provided
NM_198056.2(SCN5A):c.1127G>A (p.Arg376His)	SCN5A	3	38606682	Brugada syndrome;not provided
NM_198056.2(SCN5A):c.2632C>T (p.Arg878Cys)	SCN5A	3	38585846	Brugada syndrome;not provided
NM_198056.2(SCN5A):c.311G>A (p.Arg104Gln)	SCN5A	3	38630392	Brugada syndrome;not provided
NM_198056.2(SCN5A):c.3995C>T (p.Pro1332Leu)	SCN5A	3	38560397	Brugada syndrome;not provided
NM_198056.2(SCN5A):c.4057G>A (p.Val1353Met)	SCN5A	3	38560335	Brugada syndrome;not provided
NM_198056.2(SCN5A):c.4217G>A (p.Gly1406Glu)	SCN5A	3	38560175	Brugada syndrome;not provided
NM_198056.2(SCN5A):c.5227G>A (p.Gly1743Arg)	SCN5A	3	38551145	Brugada syndrome;not provided
NM_198056.2(SCN5A):c.2575C>T (p.Gln859Ter)	SCN5A	3	38585903	Brugada syndrome;not provided
NM_198056.2(SCN5A):c.3352C>T (p.Gln1118Ter)	SCN5A	3	38579372	Brugada syndrome;not provided
NM_000088.3(COL1A1):c.934C>T (p.Arg312Cys)	COL1A1	17	50196337	Bruising susceptibility;Ehlers-Danlos syndrome, classic type;Fragile skin;Joint hypermobility
NM_000421.3(KRT10):c.467G>A (p.Arg156His)	KRT10	17	40822119	Bullous ichthyosiform erythroderma;not provided
NM_000421.3(KRT10):c.466C>T (p.Arg156Cys)	KRT10	17	40822120	Bullous ichthyosiform erythroderma;not provided
NM_000421.3(KRT10):c.1300C>T (p.Gln434Ter)	KRT10	17	40819590	Bullous ichthyosiform erythroderma;not provided
NM_006017.2(PROM1):c.1117C>T (p.Arg373Cys)	PROM1	4	16013299	Bull's eye macular dystrophy;Cone-rod dystrophy 12;Stargardt disease 4;not provided
NM_015506.2(MMACHC):c.482G>A (p.Arg161Gln)	MMACHC	1	45508848	Bull's eye maculopathy;Methylmalonic acidemia with homocystinuria;not provided
NM_000350.2(ABCA4):c.2041C>T (p.Arg681Ter)	ABCA4	1	94060656	Bull's eye maculopathy;Stargardt disease 1;not provided
NM_002467.4(MYC):c.214C>T (p.Pro72Ser)	MYC	8	127738431	Burkitt lymphoma
NM_006701.4(TXNL4A):c.37C>T (p.Gln13Ter)	TXNL4A	18	79988356	Burn-McKeown syndrome
NM_000578.3(SLC11A1):c.1627G>A (p.Asp543Asn)	SLC11A1	2	218395009	Buruli ulcer, susceptibility to
NM_000055.2(BCHE):c.812C>T (p.Thr271Met)	BCHE	3	165830222	Butyrylcholinesterase deficiency, fluoride 1
NM_198196.2(CD96):c.839C>T (p.Thr280Met)	CD96	3	111585362	C syndrome
NM_015991.3(C1QA):c.622C>T (p.Gln208Ter)	C1QA	1	22639291	C1q deficiency
NM_003282.3(TNNI2):c.466C>T (p.Arg156Ter)	TNNI2	11	1841468	Calcaneovalgus deformity;Congenital finger flexion contractures;Distal arthrogryposis;Distal arthrogryposis type 2B;Ulnar deviation of the wrist;not provided
NM_002526.3(NT5E):c.1073G>A (p.Cys358Tyr)	NT5E	6	85487458	Calcification of joints and arteries
NM_134425.2(SLC26A1):c.554C>T (p.Thr185Met)	SLC26A1	4	991150	Calcium oxalate urolithiasis
NM_022042.3(SLC26A1):c.1073C>T (p.Ser358Leu)	SLC26A1	4	989866	Calcium oxalate urolithiasis

NM_004006.2(DMD):c.1637G>A (p.Trp546Ter)	DMD	X	32573812	Calf muscle hypertrophy;EMG abnormality;EMG: myopathic abnormalities;Motor delay;Muscle cramps;Muscle weakness
NM_000346.3(SOX9):c.472G>A (p.Ala158Thr)	SOX9	17	72122759	Campomelic dysplasia with autosomal sex reversal
NM_000142.4(FGFR3):c.1862G>A (p.Arg621His)	FGFR3	4	1806076	Camptodactyly, tall stature, and hearing loss syndrome
NM_000346.3(SOX9):c.1249C>T (p.Gln417Ter)	SOX9	17	72124106	Camptomelic dysplasia
NM_000049.2(ASPA):c.212G>A (p.Arg71His)	ASPA	17	3476371	Canavan disease, mild;Spongy degeneration of central nervous system
NM_213647.2(FGFR4):c.1162G>A (p.Gly388Arg)	FGFR4	5	177093242	Cancer progression and tumor cell motility
NM_052813.4(CARD9):c.883C>T (p.Gln295Ter)	CARD9	9	136370362	Candidiasis, familial, 2
NM_052813.4(CARD9):c.214G>A (p.Gly72Ser)	CARD9	9	136371432	Candidiasis, familial, 2
NM_052813.4(CARD9):c.865C>T (p.Gln289Ter)	CARD9	9	136370380	Candidiasis, familial, 2
NM_052813.4(CARD9):c.301C>T (p.Arg101Cys)	CARD9	9	136371345	Candidiasis, familial, 2
NM_014339.6(IL17RA):c.850C>T (p.Gln284Ter)	IL17RA	22	17104729	Candidiasis, familial, 5
NM_014339.6(IL17RA):c.1159G>A (p.Asp387Asn)	IL17RA	22	17108378	Candidiasis, familial, 5
NM_014339.6(IL17RA):c.196C>T (p.Arg66Ter)	IL17RA	22	17097829	Candidiasis, familial, 5
NM_052872.3(IL17F):c.284C>T (p.Ser95Leu)	IL17F	6	52237139	Candidiasis, familial, 6
NM_147686.3(TRAF3IP2):c.1580C>T (p.Thr527Ile)	TRAF3IP2	6	111559523	Candidiasis, familial, 8
NM_153461.3(IL17RC):c.412C>T (p.Gln138Ter)	IL17RC	3	9917994	Candidiasis, familial, 9
NM_153461.3(IL17RC):c.1126C>T (p.Arg376Ter)	IL17RC	3	9928340	Candidiasis, familial, 9
NM_153461.3(IL17RC):c.1132C>T (p.Arg378Ter)	IL17RC	3	9928346	Candidiasis, familial, 9
NM_152263.3(TPM3):c.503G>A (p.Arg168His)	TPM3	1	154172971	Cap myopathy 1;Congenital myopathy with fiber type disproportion;Nemaline myopathy 1;not provided
NM_152263.3(TPM3):c.502C>T (p.Arg168Cys)	TPM3	1	154172972	Cap myopathy 1;Congenital myopathy with fiber type disproportion;not provided
NM_005465.4(AKT3):c.1393C>T (p.Arg465Trp)	AKT3	1	243505296	Capillary hemangiomas;Global developmental delay;Macrocephaly;Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2;Polymicrogyria
NM_002890.2(RASA1):c.853C>T (p.Arg285Ter)	RASA1	5	87333291	Capillary malformation without arteriovenous malformation
NM_002890.2(RASA1):c.1619G>A (p.Cys540Tyr)	RASA1	5	87369821	Capillary malformation-arteriovenous malformation
NM_002072.4(GNAQ):c.548G>A (p.Arg183Gln)	GNAQ	9	77797577	Capillary malformations, congenital;Sturge-Weber syndrome
NM_000070.2(CAPN3):c.1469G>A (p.Arg490Gln)	CAPN3	15	42401755	CAPN3-Related Disorders;Limb-girdle muscular dystrophy, type 2A
NM_000303.2(PMM2):c.26G>A (p.Cys9Tyr)	PMM2	16	8797908	Carbohydrate-deficient glycoprotein syndrome type I
NM_000303.2(PMM2):c.710C>T (p.Thr237Met)	PMM2	16	8847794	Carbohydrate-deficient glycoprotein syndrome type I
NM_000303.2(PMM2):c.442G>A (p.Asp148Asn)	PMM2	16	8811173	Carbohydrate-deficient glycoprotein syndrome type I
NM_000303.2(PMM2):c.255+1G>A	PMM2	16	8804844	Carbohydrate-deficient glycoprotein syndrome type I
NM_000303.2(PMM2):c.422G>A (p.Arg141His)	PMM2	16	8811153	Carbohydrate-deficient glycoprotein syndrome type I;Congenital disorder of glycosylation;not provided
NM_000303.2(PMM2):c.385G>A (p.Val129Met)	PMM2	16	8811116	Carbohydrate-deficient glycoprotein syndrome type I;not provided
NM_000303.2(PMM2):c.484C>T (p.Arg162Trp)	PMM2	16	8811674	Carbohydrate-deficient glycoprotein syndrome type I;not provided
NM_000303.2(PMM2):c.691G>A (p.Val231Met)	PMM2	16	8847775	Carbohydrate-deficient glycoprotein syndrome type I;not provided
NM_000303.2(PMM2):c.338C>T (p.Pro113Leu)	PMM2	16	8806398	Carbohydrate-deficient glycoprotein syndrome type I;not provided
NM_000303.2(PMM2):c.415G>A (p.Glu139Lys)	PMM2	16	8811146	Carbohydrate-deficient glycoprotein syndrome type I;not provided
NM_002408.3(MGAT2):c.869C>T (p.Ser290Phe)	MGAT2	14	49622137	Carbohydrate-deficient glycoprotein syndrome type II
NM_001738.4(CA1):c.740G>A (p.Arg247His)	CA1	8	85328606	Carbonic anhydrase I deficiency
NM_001739.1(CA5A):c.555G>A (p.Lys185=)	CA5A	16	87902425	Carbonic anhydrase VA deficiency, hyperammonemia due to;not provided
NM_001025195.1(CES1):c.428G>A (p.Gly143Glu)	CES1	16	55823661	Carboxylesterase 1 deficiency
NM_006207.2(PDGFRL):c.67C>T (p.His23Tyr)	PDGFRL	8	17589479	Carcinoma of colon
NM_001429.3(EP300):c.1738C>T (p.Arg580Ter)	EP300	22	41137768	Carcinoma of colon

NM_138712.3(PPARG):c.863G>A (p.Arg288His)	PPARG	3	12416831	Carcinoma of colon
NM_002843.3(PTPRJ):c.640C>T (p.Arg214Cys)	PTPRJ	11	48123636	Carcinoma of colon
NM_001085377.1(MCC):c.2663C>T (p.Ala888Val)	MCC	5	113043623	Carcinoma of colon
NM_001085377.1(MCC):c.2087G>A (p.Arg696Gln)	MCC	5	113064110	Carcinoma of colon
NM_000142.4(FGFR3):c.964G>A (p.Glu322Lys)	FGFR3	4	1803725	Carcinoma of colon
NM_004333.4(BRAF):c.1391G>A (p.Gly464Glu)	BRAF	7	140781617	Carcinoma of colon;Chronic lymphocytic leukemia;Neoplasm;Rasopathy;not provided
NM_000038.5(APC):c.4012C>T (p.Gln1338Ter)	APC	5	112839606	Carcinoma of colon;Colorectal Neoplasms;Familial adenomatous polyposis 1
NM_001128425.1(MUTYH):c.1187G>A (p.Gly396Asp)	MUTYH	1	45331556	Carcinoma of colon;Endometrial carcinoma;Hereditary cancer-predisposing syndrome;MYH-associated polyposis;Neoplasm of stomach;not provided;not specified
NM_001128425.1(MUTYH):c.55C>T (p.Arg19Ter)	MUTYH	1	45334493	Carcinoma of colon;Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.1214C>T (p.Pro405Leu)	MUTYH	1	45331529	Carcinoma of colon;Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.739C>T (p.Arg247Ter)	MUTYH	1	45332440	Carcinoma of colon;Hereditary cancer-predisposing syndrome;not provided
NM_001128425.1(MUTYH):c.1171C>T (p.Gln391Ter)	MUTYH	1	45331676	Carcinoma of colon;MYH-associated polyposis;not provided
NM_001127500.2(MET):c.3328G>A (p.Val1110Ile)	MET	7	116777403	Carcinoma;Hereditary cancer-predisposing syndrome;Neoplasm
NM_001127500.2(MET):c.3736G>A (p.Asp1246Asn)	MET	7	116783353	Carcinoma;Renal cell carcinoma, papillary, 1
NM_199037.4(SCN1B):c.537G>A (p.Trp179Ter)	SCN1B	19	35033828	Cardiac conduction defect, nonspecific
NM_015978.2(TNNI3K):c.1577G>A (p.Gly526Asp)	TNNI3K	1	74369495	Cardiac conduction disease with or without dilated cardiomyopathy
NM_000368.4(TSC1):c.2356C>T (p.Arg786Ter)	TSC1	9	132902640	Cardiac rhabdomyoma;Hamartoma;Seizures;Tuberous sclerosis 1;Tuberous sclerosis syndrome
NM_001110556.1(FLNA):c.862G>A (p.Gly288Arg)	FLNA	X	154367403	Cardiac valvular dysplasia, X-linked
NM_001110556.1(FLNA):c.1923C>T (p.Gly641=)	FLNA	X	154364625	Cardiac valvular dysplasia, X-linked;Periventricular nodular heterotopia 1
NM_001169109.1(SCO2):c.674C>T (p.Ser225Phe)	SCO2	22	50523738	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
NM_001169109.1(SCO2):c.511C>T (p.Arg171Trp)	SCO2	22	50523901	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
NM_001169109.1(SCO2):c.268C>T (p.Arg90Ter)	SCO2	22	50524144	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
NM_001169109.1(SCO2):c.398G>A (p.Cys133Tyr)	SCO2	22	50524014	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
NM_001169109.1(SCO2):c.107G>A (p.Trp36Ter)	SCO2	22	50524305	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
NM_004376.6(COX15):c.649C>T (p.Arg217Trp)	COX15	10	99724057	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2;Congenital myasthenic syndrome, acetazolamide-responsive
NM_001169109.1(SCO2):c.157C>T (p.Gln53Ter)	SCO2	22	50524255	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency;Myopia 6
NM_005138.2(SCO2):c.418G>A (p.Glu140Lys)	SCO2	22	50523994	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency;Myopia 6;not provided
NM_004333.4(BRAF):c.1720C>T (p.His574Tyr)	BRAF	7	140754208	Cardio-facio-cutaneous syndrome
NM_004333.4(BRAF):c.1501G>A (p.Glu501Lys)	BRAF	7	140778007	Cardiofaciocutaneous syndrome 1;Noonan syndrome 7;Rasopathy;not provided
NM_030662.3(MAP2K2):c.395G>A (p.Gly132Asp)	MAP2K2	19	4110564	Cardiofaciocutaneous syndrome 4;Rasopathy;not provided
NM_002755.3(MAP2K1):c.199G>A (p.Asp67Asn)	MAP2K1	15	66435145	Cardio-facio-cutaneous syndrome;Cardiofaciocutaneous syndrome 3;Malignant melanoma;Non-small cell lung cancer;Rasopathy;not provided
NM_002755.3(MAP2K1):c.371C>T (p.Pro124Leu)	MAP2K1	15	66436825	Cardio-facio-cutaneous syndrome;Colorectal Neoplasms;Malignant melanoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Transitional cell carcinoma of the bladder
m.8363G>A	MT-TK	MT	8363	Cardiomyopathy and Deafness;Leigh syndrome;Myoclonus with epilepsy with ragged red fibers
NM_004415.3(DSP):c.3799C>T (p.Arg1267Ter)	DSP	6	7579989	Cardiomyopathy dilated with woolly hair and keratoderma;Long QT syndrome;Primary dilated cardiomyopathy
m.3303C>T	MT-TL1	MT	3303	Cardiomyopathy with or without skeletal myopathy
m.8529G>A	MT-ATP8	MT	8529	Cardiomyopathy, apical hypertrophic, and neuropathy
NM_002880.3(RAF1):c.1922C>T (p.Thr641Met)	RAF1	3	12584539	Cardiomyopathy, dilated, 1NN
NM_002880.3(RAF1):c.709G>A (p.Ala237Thr)	RAF1	3	12604261	Cardiomyopathy, dilated, 1NN
NM_004415.3(DSP):c.1691C>T (p.Thr564Ile)	DSP	6	7570553	Cardiomyopathy, dilated, with woolly hair, keratoderma, and tooth agenesis

NM_004415.3(DSP):c.1790C>T (p.Ser597Leu)	DSP	6	7571471	Cardiomyopathy, dilated, with woolly hair, keratoderma, and tooth agenesis;not provided
NM_032578.3(MYPN):c.1585C>T (p.Gln529Ter)	MYPN	10	68165803	Cardiomyopathy, familial restrictive, 4;not provided
NM_001458.4(FLNC):c.4871C>T (p.Ser1624Leu)	FLNC	7	128848926	Cardiomyopathy, familial restrictive, 5
NM_033118.3(MYLK2):c.260C>T (p.Ala87Val)	MYLK2	20	31820333	Cardiomyopathy, hypertrophic, midventricular, digenic
m.12192G>A	MT-TH	MT	12192	Cardiomyopathy, hypertrophic, mitochondrial;Cardiomyopathy, idiopathic dilated, mitochondrial
NM_000363.4(TNNI3):c.508C>T (p.Arg170Trp)	TNNI3	19	55154071	Cardiomyopathy, restrictive;Familial restrictive cardiomyopathy;Hypertrophic cardiomyopathy;not provided
NM_001103.3(ACTN2):c.355G>A (p.Ala119Thr)	ACTN2	1	236719007	Cardiomyopathy;Dilated cardiomyopathy 1AA;Familial hypertrophic cardiomyopathy 23
NM_000257.3(MYH7):c.1573G>A (p.Glu525Lys)	MYH7	14	23428505	Cardiomyopathy;Familial cardiomyopathy;Hypertrophic cardiomyopathy;Left ventricular noncompaction cardiomyopathy
NM_000363.4(TNNI3):c.575G>A (p.Arg192His)	TNNI3	19	55151892	Cardiomyopathy;Familial restrictive cardiomyopathy 1;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.4498C>T (p.Arg1500Trp)	MYH7	14	23417174	Cardiomyopathy;Hypertrophic cardiomyopathy
NM_000257.3(MYH7):c.2302G>A (p.Gly768Arg)	MYH7	14	23425403	Cardiomyopathy;Hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.1351+1G>A	MYBPC3	11	47343020	Cardiomyopathy;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy
NM_001001430.2(TNNT2):c.421C>T (p.Arg141Trp)	TNNT2	1	201364336	Cardiomyopathy;Left ventricular noncompaction 6;Primary dilated cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000363.4(TNNI3):c.509G>A (p.Arg170Gln)	TNNI3	19	55154070	Cardiomyopathy;not provided
NM_000238.3(KCNH2):c.1557+1G>A	KCNH2	7	150952424	Cardiovascular phenotype
NM_004281.3(BAG3):c.481C>T (p.Gln161Ter)	BAG3	10	119670151	Cardiovascular phenotype
NM_000218.2(KCNQ1):c.564G>A (p.Trp188Ter)	KCNQ1	11	2570714	Cardiovascular phenotype
NM_001035.2(RYR2):c.14311G>A (p.Val4771Ile)	RYR2	1	237808913	Cardiovascular phenotype;Catecholaminergic polymorphic ventricular tachycardia;not provided
NM_170707.3(LMNA):c.1621C>T (p.Arg541Cys)	LMNA	1	156137666	Cardiovascular phenotype;Charcot-Marie-Tooth disease, type 2;Dilated cardiomyopathy 1A;Primary dilated cardiomyopathy;not provided
NM_170707.3(LMNA):c.1057C>T (p.Gln353Ter)	LMNA	1	156136021	Cardiovascular phenotype;Charcot-Marie-Tooth disease, type 2;not provided
NM_000219.5(KCNE1):c.226G>A (p.Asp76Asn)	KCNE1	21	34449409	Cardiovascular phenotype;Congenital long QT syndrome;Jervell and Lange-Nielsen syndrome 1;Jervell and Lange-Nielsen syndrome 2;Long QT syndrome;Long QT syndrome 5;not provided
NM_000238.3(KCNH2):c.2464G>A (p.Val822Met)	KCNH2	7	150948984	Cardiovascular phenotype;Congenital long QT syndrome;Long QT syndrome 2
NM_000218.2(KCNQ1):c.613G>A (p.Val205Met)	KCNQ1	11	2571333	Cardiovascular phenotype;Congenital long QT syndrome;Long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype;not provided
NM_000218.2(KCNQ1):c.1781G>A (p.Arg594Gln)	KCNQ1	11	2778024	Cardiovascular phenotype;Congenital long QT syndrome;Long QT syndrome;not provided
NM_000218.2(KCNQ1):c.965C>T (p.Thr322Met)	KCNQ1	11	2583478	Cardiovascular phenotype;Congenital long QT syndrome;Long QT syndrome;not provided
NM_000238.3(KCNH2):c.1838C>T (p.Thr613Met)	KCNH2	7	150951555	Cardiovascular phenotype;Congenital long QT syndrome;not provided
NM_198056.2(SCN5A):c.4877G>A (p.Arg1626His)	SCN5A	3	38551495	Cardiovascular phenotype;Congenital long QT syndrome;not provided
NM_003319.4(TTN):c.57895C>T (p.Arg19299Ter)	TTN	2	178561042	Cardiovascular phenotype;Dilated cardiomyopathy 1G;Primary dilated cardiomyopathy
NM_004281.3(BAG3):c.367C>T (p.Arg123Ter)	BAG3	10	119670037	Cardiovascular phenotype;Dilated cardiomyopathy 1HH;Primary dilated cardiomyopathy;not provided
NM_000257.3(MYH7):c.1988G>A (p.Arg663His)	MYH7	14	23426833	Cardiovascular phenotype;Dilated cardiomyopathy 1S;Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Myopathy, distal, 1;Myopathy, myosin storage, autosomal recessive;Myosin storage myopathy;Primary familial hypertrophic cardiomyopathy;Scapuloperoneal myopathy, MYH7-related;not provided
NM_000169.2(GLA):c.902G>A (p.Arg301Gln)	GLA	X	101398467	Cardiovascular phenotype;Fabry disease;Fabry disease, cardiac variant
NM_000257.3(MYH7):c.1816G>A (p.Val606Met)	MYH7	14	23427657	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.2770G>A (p.Glu924Lys)	MYH7	14	23424059	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided

NM_000257.3(MYH7):c.2167C>T (p.Arg723Cys)	MYH7	14	23425814	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.2156G>A (p.Arg719Gln)	MYH7	14	23425970	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.2155C>T (p.Arg719Trp)	MYH7	14	23425971	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 1;Primary familial hypertrophic cardiomyopathy;not provided
NM_001001430.2(TNNT2):c.388C>T (p.Arg130Cys)	TNNT2	1	201364369	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 2;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.2308G>A (p.Asp770Asn)	MYBPC3	11	47338520	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.2827C>T (p.Arg943Ter)	MYBPC3	11	47335120	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.1505G>A (p.Arg502Gln)	MYBPC3	11	47342697	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.772G>A (p.Glu258Lys)	MYBPC3	11	47348424	Cardiovascular phenotype;Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.772+1G>A	MYBPC3	11	47348423	Cardiovascular phenotype;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.821+1G>A	MYBPC3	11	47347856	Cardiovascular phenotype;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.2221G>A (p.Gly741Arg)	MYH7	14	23425760	Cardiovascular phenotype;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_002834.4(PTPN11):c.1403C>T (p.Thr468Met)	PTPN11	12	112488466	Cardiovascular phenotype;LEOPARD syndrome 1;Noonan syndrome;Noonan syndrome 1;Noonan syndrome with multiple lentigines;Rasopathy;not provided
NM_000363.4(TNNI3):c.523C>T (p.Gln175Ter)	TNNI3	19	55154056	Cardiovascular phenotype;not provided
NM_133378.4(TTN):c.81313C>T (p.Arg27105Ter)	TTN	2	178554094	Cardiovascular phenotype;not provided
NM_198056.2(SCN5A):c.4885C>T (p.Arg1629Ter)	SCN5A	3	38551487	Cardiovascular phenotype;not provided
NM_000256.3(MYBPC3):c.655-1G>A	MYBPC3	11	47348542	Cardiovascular phenotype;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.2788G>A (p.Glu930Lys)	MYH7	14	23424041	Cardiovascular phenotype;Primary familial hypertrophic cardiomyopathy;not provided
NM_024027.4(COLEC11):c.610G>A (p.Gly204Ser)	COLEC11	2	3643912	Carnevale syndrome
NM_002472.2(MYH8):c.2021G>A (p.Arg674Gln)	MYH8	17	10406924	Carney complex variant;Hecht syndrome;not provided
NM_212472.2(PRKAR1A):c.109C>T (p.Gln37Ter)	PRKAR1A	17	68515508	Carney complex, type 1
NM_212472.2(PRKAR1A):c.124C>T (p.Arg42Ter)	PRKAR1A	17	68515523	Carney complex, type 1
NM_212472.2(PRKAR1A):c.286C>T (p.Arg96Ter)	PRKAR1A	17	68522864	Carney complex, type 1
NM_212472.2(PRKAR1A):c.682C>T (p.Arg228Ter)	PRKAR1A	17	68525886	Carney complex, type 1
NM_212472.2(PRKAR1A):c.82C>T (p.Gln28Ter)	PRKAR1A	17	68515481	Carney complex, type 1
NM_003001.3(SDHC):c.405+1G>A	SDHC	1	161356841	Carney triad;Gastrointestinal stromal tumor;Paraganglioma and gastric stromal sarcoma
NM_004168.3(SDHA):c.91C>T (p.Arg31Ter)	SDHA	5	223509	Carney triad;Hereditary cancer-predisposing syndrome;Mitochondrial complex II deficiency;Paragangliomas 5;not provided
NM_000387.5(SLC25A20):c.496C>T (p.Arg166Ter)	SLC25A20	3	48862581	Carnitine acylcarnitine translocase deficiency
NM_000387.5(SLC25A20):c.576G>A (p.Trp192Ter)	SLC25A20	3	48859587	Carnitine acylcarnitine translocase deficiency
NM_001876.3(CPT1A):c.1241C>T (p.Ala414Val)	CPT1A	11	68781882	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.1876-1G>A (p.Val626_Glu676del)	CPT1A	11	68761688	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.2126G>A (p.Gly709Glu)	CPT1A	11	68760241	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.2129G>A (p.Gly710Glu)	CPT1A	11	68760238	Carnitine palmitoyltransferase I deficiency

NM_001876.3(CPT1A):c.1069C>T (p.Arg357Trp)	CPT1A	11	68784909	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.1339C>T (p.Arg447Ter)	CPT1A	11	68781784	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.1425G>A (p.Trp475Ter)	CPT1A	11	68780673	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.2156G>A (p.Gly719Asp)	CPT1A	11	68759648	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.478C>T (p.Arg160Ter)	CPT1A	11	68804077	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.941C>T (p.Thr314Ile)	CPT1A	11	68793341	Carnitine palmitoyltransferase I deficiency
NM_001876.3(CPT1A):c.298C>T (p.Gln100Ter)	CPT1A	11	68807622	Carnitine palmitoyltransferase I deficiency;not provided
NM_001876.3(CPT1A):c.1436C>T (p.Pro479Leu)	CPT1A	11	68780662	Carnitine palmitoyltransferase I deficiency;not provided
NM_001031847.2(CPT1A):c.281+1G>A	CPT1A	11	68812436	Carnitine palmitoyltransferase I deficiency;not provided
NM_000098.2(CPT2):c.1145G>A (p.Arg382Lys)	CPT2	1	53210819	Carnitine palmitoyltransferase II deficiency
NM_000098.2(CPT2):c.452G>A (p.Arg151Gln)	CPT2	1	53210126	Carnitine palmitoyltransferase II deficiency
NM_000098.2(CPT2):c.1646G>A (p.Gly549Asp)	CPT2	1	53213264	Carnitine palmitoyltransferase II deficiency
NM_000098.2(CPT2):c.1657G>A (p.Asp553Asn)	CPT2	1	53213275	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED
NM_000098.2(CPT2):c.1507C>T (p.Arg503Cys)	CPT2	1	53211181	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED;Carnitine palmitoyltransferase II deficiency
NM_000098.2(CPT2):c.886C>T (p.Arg296Ter)	CPT2	1	53210560	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED;Carnitine palmitoyltransferase II deficiency, infantile
NM_000098.2(CPT2):c.370C>T (p.Arg124Ter)	CPT2	1	53210044	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED;Carnitine palmitoyltransferase II deficiency, infantile;Carnitine palmitoyltransferase II deficiency, lethal neonatal;not provided
NM_000098.2(CPT2):c.1891C>T (p.Arg631Cys)	CPT2	1	53213509	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED;Carnitine palmitoyltransferase II deficiency;Carnitine palmitoyltransferase II deficiency, infantile
NM_000098.2(CPT2):c.338C>T (p.Ser113Leu)	CPT2	1	53202427	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED;Carnitine palmitoyltransferase II deficiency;Carnitine palmitoyltransferase II deficiency, lethal neonatal;not provided
NM_000098.2(CPT2):c.520G>A (p.Glu174Lys)	CPT2	1	53210194	Carnitine palmitoyltransferase II deficiency;Carnitine palmitoyltransferase II deficiency, infantile
NM_001410.2(MEGF8):c.4496G>A (p.Arg1499His)	MEGF8	19	42356848	Carpenter syndrome 2
NM_001410.2(MEGF8):c.1342C>T (p.Arg448Ter)	MEGF8	19	42336904	Carpenter syndrome 2
NM_001228.4(CASP8):c.793C>T (p.Arg265Trp)	CASP8	2	201276908	Caspase-8 deficiency
NM_005267.4(GJA8):c.262C>T (p.Pro88Ser)	GJA8	1	147908217	Cataract 1
NM_005267.4(GJA8):c.142G>A (p.Glu48Lys)	GJA8	1	147908097	Cataract 1
NM_005267.4(GJA8):c.593G>A (p.Arg198Gln)	GJA8	1	147908548	Cataract 1
NM_005267.4(GJA8):c.139G>A (p.Asp47Asn)	GJA8	1	147908094	Cataract 1
NM_005267.4(GJA8):c.566C>T (p.Pro189Leu)	GJA8	1	147908521	Cataract 1
NM_001885.2(CRYAB):c.418G>A (p.Asp140Asn)	CRYAB	11	111908874	Cataract 16, multiple types
NM_001885.2(CRYAB):c.58C>T (p.Pro20Ser)	CRYAB	11	111911667	Cataract 16, multiple types
NM_001885.2(CRYAB):c.166C>T (p.Arg56Trp)	CRYAB	11	111911559	Cataract 16, multiple types
NM_030657.3(LIM2):c.587G>A (p.Gly196Glu)	LIM2	19	51380262	Cataract 19, multiple types
NM_000496.2(CRYBB2):c.463C>T (p.Gln155Ter)	CRYBB2	22	25231617	Cataract 3, multiple types
NM_003380.3(VIM):c.451G>A (p.Glu151Lys)	VIM	10	17229873	Cataract 30;not provided
NM_012186.2(FOXE3):c.307G>A (p.Glu103Lys)	FOXE3	1	47416622	Cataract 34, multiple types
NM_006891.3(CRYGD):c.43C>T (p.Arg15Cys)	CRYGD	2	208124321	Cataract 4
NM_006891.3(CRYGD):c.176G>A (p.Arg59His)	CRYGD	2	208124188	Cataract 4
NM_006891.3(CRYGD):c.470G>A (p.Trp157Ter)	CRYGD	2	208121728	Cataract 4
NM_006891.3(CRYGD):c.70C>T (p.Pro24Ser)	CRYGD	2	208124294	Cataract 4

NM_173167.3(UNC45B):c.2413C>T (p.Arg805Trp)	UNC45B	17	35183460	Cataract 43
NM_001001438.2(LSS):c.1762G>A (p.Gly588Ser)	LSS	21	46195731	Cataract 44
NM_015073.2(SIPA1L3):c.4489C>T (p.Arg1497Ter)	SIPA1L3	19	38192203	Cataract 45
NM_004431.4(EPHA2):c.2819C>T (p.Thr940Ile)	EPHA2	1	16129440	Cataract 6, multiple types
NM_000394.3(CRYAA):c.27G>A (p.Trp9Ter)	CRYAA	21	43169126	Cataract 9, autosomal recessive
NM_018238.3(AGK):c.517C>T (p.Gln173Ter)	AGK	7	141615564	Cataract and cardiomyopathy
NM_018238.3(AGK):c.841C>T (p.Arg281Ter)	AGK	7	141641362	Cataract and cardiomyopathy;not provided
NM_018238.3(AGK):c.409C>T (p.Arg137Ter)	AGK	7	141614164	Cataract and cardiomyopathy;not provided
NM_000394.3(CRYAA):c.346C>T (p.Arg116Cys)	CRYAA	21	43172104	Cataract, autosomal dominant
NM_000394.3(CRYAA):c.145C>T (p.Arg49Cys)	CRYAA	21	43169244	Cataract, autosomal dominant
NM_000394.3(CRYAA):c.292G>A (p.Gly98Arg)	CRYAA	21	43170619	Cataract, autosomal dominant
NM_000394.3(CRYAA):c.160C>T (p.Arg54Cys)	CRYAA	21	43169259	Cataract, autosomal dominant
NM_000394.3(CRYAA):c.62G>A (p.Arg21Gln)	CRYAA	21	43169161	Cataract, autosomal dominant
NM_003571.3(BFSP2):c.859C>T (p.Arg287Trp)	BFSP2	3	133450432	Cataract, autosomal dominant, multiple types 1;not provided
NM_000394.3(CRYAA):c.34C>T (p.Arg12Cys)	CRYAA	21	43169133	Cataract, autosomal dominant, multiple types, with microcornea
NM_000394.3(CRYAA):c.61C>T (p.Arg21Trp)	CRYAA	21	43169160	Cataract, autosomal dominant, multiple types, with microcornea;Congenital cataract
NM_000394.3(CRYAA):c.347G>A (p.Arg116His)	CRYAA	21	43172105	Cataract, autosomal dominant;Cataract, autosomal dominant, multiple types, with microcornea;not provided
NM_024513.3(FYCO1):c.1045C>T (p.Gln349Ter)	FYCO1	3	45968289	Cataract, autosomal recessive congenital 2
NM_024513.3(FYCO1):c.2206C>T (p.Gln736Ter)	FYCO1	3	45967128	Cataract, autosomal recessive congenital 2
NM_024513.3(FYCO1):c.1546C>T (p.Gln516Ter)	FYCO1	3	45967788	Cataract, autosomal recessive congenital 2
NM_005029.3(PITX3):c.38G>A (p.Ser13Asn)	PITX3	10	102232043	Cataract, congenital
NM_020989.3(CRYGC):c.470G>A (p.Trp157Ter)	CRYGC	2	208128258	Cataract, coppock-like
NM_020989.3(CRYGC):c.471G>A (p.Trp157Ter)	CRYGC	2	208128257	Cataract, coppock-like
NM_020989.3(CRYGC):c.497C>T (p.Ser166Phe)	CRYGC	2	208128231	Cataract, coppock-like;Congenital cataract
NM_213606.3(SLC16A12):c.733C>T (p.Gln245Ter)	SLC16A12	10	89438899	Cataract, juvenile, with microcornea and glucosuria
NM_176812.4(CHMP4B):c.481G>A (p.Glu161Lys)	CHMP4B	20	33851064	Cataract, posterior polar, 3
NM_001040667.2(HSF4):c.355C>T (p.Arg119Cys)	HSF4	16	67165841	Cataract, zonular
NM_018060.3(IARS2):c.2726C>T (p.Pro909Leu)	IARS2	1	220143109	Cataract;Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia;Peripheral neuropathy;growth hormone deficiency with short stature;partial sensorineural deafness
NM_001035.2(RYR2):c.6916G>A (p.Val2306Ile)	RYR2	1	237638480	Catecholaminergic polymorphic ventricular tachycardia
NM_001035.2(RYR2):c.506G>A (p.Arg169Gln)	RYR2	1	237377365	Catecholaminergic polymorphic ventricular tachycardia;not provided
NM_001232.3(CASQ2):c.923C>T (p.Pro308Leu)	CASQ2	1	115705208	Catecholaminergic polymorphic ventricular tachycardia;not provided
NM_001035.2(RYR2):c.11836G>A (p.Gly3946Ser)	RYR2	1	237778726	Catecholaminergic polymorphic ventricular tachycardia;not provided
NM_001035.2(RYR2):c.1258C>T (p.Arg420Trp)	RYR2	1	237445488	Catecholaminergic polymorphic ventricular tachycardia;Ventricular fibrillation;not provided
NM_001035.2(RYR2):c.6737C>T (p.Ser2246Leu)	RYR2	1	237634937	Catecholaminergic polymorphic ventricular tachycardia;Ventricular tachycardia, catecholaminergic polymorphic, 1;not provided
NM_138959.2(VANGL1):c.715G>A (p.Val239Ile)	VANGL1	1	115664171	Caudal regression syndrome;not provided
NM_203330.2(CD59):c.266G>A (p.Cys89Tyr)	CD59	11	33710247	Cd59 deficiency
NM_001768.6(CD8A):c.331G>A (p.Gly111Ser)	CD8A	2	86790400	Cd8 deficiency, familial
NM_022124.5(CDH23):c.5237G>A (p.Arg1746Gln)	CDH23	10	71779316	CDH23-Related Disorders;Usher syndrome, type 1D;not provided
NM_000540.2(RYR1):c.14545G>A (p.Val4849Ile)	RYR1	19	38580403	Central core disease, autosomal recessive;Minicore myopathy with external ophthalmoplegia;not provided
NM_000540.2(RYR1):c.10579C>T (p.Pro3527Ser)	RYR1	19	38525455	Central core disease, autosomal recessive;not provided

NM_000540.2(RYR1):c.12612G>A (p.Trp4204Ter)	RYR1	19	38561442	Central core disease;Malignant hyperthermia, susceptibility to, 1
NM_000540.2(RYR1):c.487C>T (p.Arg163Cys)	RYR1	19	38444211	Central core disease;Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.7373G>A (p.Arg2458His)	RYR1	19	38500655	Central core disease;Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.6487C>T (p.Arg2163Cys)	RYR1	19	38494564	Central core disease;Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.6488G>A (p.Arg2163His)	RYR1	19	38494565	Central core disease;Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.7522C>T (p.Arg2508Cys)	RYR1	19	38500898	Central core disease;Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.14126C>T (p.Thr4709Met)	RYR1	19	38573304	Central core disease;Minicore myopathy with external ophthalmoplegia;not provided
NM_000540.2(RYR1):c.14582G>A (p.Arg4861His)	RYR1	19	38580440	Central core disease;Neuromuscular disease, congenital, with uniform type 1 fiber;not provided
NM_000540.2(RYR1):c.7304G>A (p.Arg2435His)	RYR1	19	38499997	Central core disease;not provided
NM_000540.2(RYR1):c.4729G>A (p.Ala1577Thr)	RYR1	19	38483311	Central core disease;not provided
NM_000540.2(RYR1):c.14537C>T (p.Ala4846Val)	RYR1	19	38580395	Central core disease;not provided
NM_000540.2(RYR1):c.178G>A (p.Asp60Asn)	RYR1	19	38442361	Central core disease;not provided
NM_000540.2(RYR1):c.13900G>A (p.Glu4634Lys)	RYR1	19	38572172	Central core disease;not provided
NM_000540.2(RYR1):c.1534G>A (p.Glu512Lys)	RYR1	19	38435328	Central core disease;not provided
NM_000540.2(RYR1):c.644G>A (p.Gly215Glu)	RYR1	19	38446484	Central core disease;not provided
NM_000540.2(RYR1):c.13913G>A (p.Gly4638Asp)	RYR1	19	38572185	Central core disease;not provided
NM_000540.2(RYR1):c.13912G>A (p.Gly4638Ser)	RYR1	19	38572184	Central core disease;not provided
NM_000540.2(RYR1):c.14696G>A (p.Gly4899Glu)	RYR1	19	38584992	Central core disease;not provided
NM_000540.2(RYR1):c.14659C>T (p.His4887Tyr)	RYR1	19	38584955	Central core disease;not provided
NM_000540.2(RYR1):c.14440C>T (p.Leu4814Phe)	RYR1	19	38580057	Central core disease;not provided
NM_000540.2(RYR1):c.7354C>T (p.Arg2452Trp)	RYR1	19	38500636	Central core disease;not provided
NM_000540.2(RYR1):c.7361G>A (p.Arg2454His)	RYR1	19	38500643	Central core disease;not provided
NM_000540.2(RYR1):c.8816G>A (p.Arg2939Lys)	RYR1	19	38506952	Central core disease;not provided
NM_000540.2(RYR1):c.14473C>T (p.Arg4825Cys)	RYR1	19	38580090	Central core disease;not provided
NM_000540.2(RYR1):c.14581C>T (p.Arg4861Cys)	RYR1	19	38580439	Central core disease;not provided
NM_000540.2(RYR1):c.14678G>A (p.Arg4893Gln)	RYR1	19	38584974	Central core disease;not provided
NM_000540.2(RYR1):c.14677C>T (p.Arg4893Trp)	RYR1	19	38584973	Central core disease;not provided
NM_000540.2(RYR1):c.1280C>T (p.Ser427Leu)	RYR1	19	38452854	Central core disease;not provided
NM_000540.2(RYR1):c.13910C>T (p.Thr4637Ile)	RYR1	19	38572182	Central core disease;not provided
NM_006892.3(DNMT3B):c.2452G>A (p.Val818Met)	DNMT3B	20	32807793	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
NM_006892.3(DNMT3B):c.1987G>A (p.Gly663Ser)	DNMT3B	20	32800916	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
NM_006892.3(DNMT3B):c.1807G>A (p.Ala603Thr)	DNMT3B	20	32800200	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
NM_175850.2(DNMT3B):c.2397-11G>A	DNMT3B	20	32807751	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
NM_006892.3(DNMT3B):c.88C>T (p.Gln30Ter)	DNMT3B	20	32780411	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
NM_006892.3(DNMT3B):c.2519G>A (p.Arg840Gln)	DNMT3B	20	32807860	Centromeric instability of chromosomes 1,9 and 16 and immunodeficiency
NM_003383.4(VLDLR):c.769C>T (p.Arg257Ter)	VLDLR	9	2643480	Cerebellar ataxia and mental retardation with quadrupedal locomotion 1;Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1
NM_002693.2(POLG):c.2557C>T (p.Arg853Trp)	POLG	15	89321777	Cerebellar ataxia infantile with progressive external ophthalmoplegia
NM_002693.2(POLG):c.2542G>A (p.Gly848Ser)	POLG	15	89321792	Cerebellar ataxia infantile with progressive external ophthalmoplegia;Mitochondrial DNA depletion syndrome 4B, MNGIE type;POLG-Related Spectrum Disorders;Progressive external ophthalmoplegia with mitochondrial DNA deletions, digenic;Progressive sclerosing poliodystrophy;not provided
NM_002693.2(POLG):c.2554C>T (p.Arg852Cys)	POLG	15	89321780	Cerebellar ataxia infantile with progressive external ophthalmoplegia;Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis;not provided
NM_001130823.2(DNMT1):c.1709C>T (p.Ala570Val)	DNMT1	19	10154709	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant
NM_003383.4(VLDLR):c.1342C>T (p.Arg448Ter)	VLDLR	9	2645603	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1

NM_001163809.1(WDR81):c.2567C>T (p.Pro856Leu)	WDR81	17	1727526	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2
NM_001163673.1(WDR81):c.388C>T (p.Arg130Ter)	WDR81	17	1731098	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2
NM_004056.5(CA8):c.710G>A (p.Arg237Gln)	CA8	8	60222677	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 3
NM_003383.4(VLDLR):c.83-1G>A	VLDLR	9	2635452	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1
NM_001519.3(BRF1):c.776C>T (p.Thr259Met)	BRF1	14	105228832	Cerebellofaciodental syndrome;cerebellar-facial-dental syndrome
NM_001519.3(BRF1):c.677C>T (p.Ser226Leu)	BRF1	14	105241282	Cerebellofaciodental syndrome;cerebellar-facial-dental syndrome
NM_001519.3(BRF1):c.667C>T (p.Arg223Trp)	BRF1	14	105241292	Cerebellofaciodental syndrome;cerebellar-facial-dental syndrome
NM_015102.4(NPHP4):c.2044C>T (p.Arg682Ter)	NPHP4	1	5904716	Cerebello-oculo-renal syndrome (nephronophthisis, oculomotor apraxia and cerebellar abnormalities);Infertility;Nephronophthisis 4
NM_000484.3(APP):c.2077G>A (p.Glu693Lys)	APP	21	25891856	Cerebral amyloid angiopathy, APP-related;not provided
NM_000484.3(APP):c.2080G>A (p.Asp694Asn)	APP	21	25891853	Cerebral amyloid angiopathy, APP-related;not provided
NM_000311.4(PRNP):c.478C>T (p.Gln160Ter)	PRNP	20	4699698	CEREBRAL AMYLOID ANGIOPATHY, PRNP-RELATED;Genetic prion diseases
NM_002775.4(HTRA1):c.973_1005del33	HTRA1	10	122507369	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2
NM_000435.2(NOTCH3):c.505C>T (p.Arg169Cys)	NOTCH3	19	15192134	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
NM_000435.2(NOTCH3):c.544C>T (p.Arg182Cys)	NOTCH3	19	15192095	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
NM_000435.2(NOTCH3):c.187G>A (p.Ala63Thr)	NOTCH3	19	15197510	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
NM_000435.2(NOTCH3):c.994C>T (p.Arg332Cys)	NOTCH3	19	15191466	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
NM_000435.2(NOTCH3):c.457C>T (p.Arg153Cys)	NOTCH3	19	15192182	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
NM_000435.2(NOTCH3):c.397C>T (p.Arg133Cys)	NOTCH3	19	15192242	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy;Recurrent subcortical infarcts
NM_002775.4(HTRA1):c.1108C>T (p.Arg370Ter)	HTRA1	10	122508758	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
NM_002775.4(HTRA1):c.904C>T (p.Arg302Ter)	HTRA1	10	122506817	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
NM_002775.4(HTRA1):c.889G>A (p.Val297Met)	HTRA1	10	122506802	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
NM_002775.4(HTRA1):c.754G>A (p.Ala252Thr)	HTRA1	10	122489603	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
NM_002775.4(HTRA1):c.883G>A (p.Gly295Arg)	HTRA1	10	122506796	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
NM_002775.4(HTRA1):c.821G>A (p.Arg274Gln)	HTRA1	10	122506734	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
NM_002775.4(HTRA1):c.961G>A (p.Ala321Thr)	HTRA1	10	122506874	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
NM_194456.1(KRIT1):c.715C>T (p.Gln239Ter)	KRIT1	7	92235417	Cerebral cavernous malformation
NM_194456.1(KRIT1):c.1363C>T (p.Gln455Ter)	KRIT1	7	92222870	Cerebral cavernous malformation;Cerebral cavernous malformations 1;not provided
NM_031443.3(CCM2):c.319C>T (p.Gln107Ter)	CCM2	7	45064493	Cerebral cavernous malformations 2;not provided
NM_016724.2(FOLR1):c.352C>T (p.Gln118Ter)	FOLR1	11	72195454	Cerebral folate deficiency
NM_016824.4(ADD3):c.1100G>A (p.Gly367Asp)	ADD3	10	110122249	Cerebral palsy;Cerebral palsy, spastic quadriplegic, 3
NM_000124.3(ERCC6):c.3862C>T (p.Arg1288Ter)	ERCC6	10	49461473	Cerebro-oculo-facio-skeletal syndrome
NM_000400.3(ERCC2):c.2041G>A (p.Asp681Asn)	ERCC2	19	45352511	Cerebrooculofacioskeletal syndrome 2
NM_001983.3(ERCC1):c.472C>T (p.Gln158Ter)	ERCC1	19	45419151	Cerebrooculofacioskeletal syndrome 4
NM_000400.3(ERCC2):c.1846C>T (p.Arg616Trp)	ERCC2	19	45352802	Cerebro-oculo-facio-skeletal syndrome;Cerebrooculofacioskeletal syndrome 2;Xeroderma pigmentosum, group D
NM_025099.5(CTC1):c.2959C>T (p.Arg987Trp)	CTC1	17	8229943	Cerebroretinal microangiopathy with calcifications and cysts
NM_025099.5(CTC1):c.2611G>A (p.Val871Met)	CTC1	17	8231334	Cerebroretinal microangiopathy with calcifications and cysts
NM_025099.5(CTC1):c.775G>A (p.Val259Met)	CTC1	17	8237392	Cerebroretinal microangiopathy with calcifications and cysts
NM_025099.5(CTC1):c.2518C>T (p.Arg840Trp)	CTC1	17	8231427	Cerebroretinal microangiopathy with calcifications and cysts
NM_025099.5(CTC1):c.3583C>T (p.Arg1195Ter)	CTC1	17	8228251	Cerebroretinal microangiopathy with calcifications and cysts
NM_025099.5(CTC1):c.680C>T (p.Ala227Val)	CTC1	17	8237487	Cerebroretinal microangiopathy with calcifications and cysts
NM_025099.5(CTC1):c.859C>T (p.Arg287Ter)	CTC1	17	8236276	Cerebroretinal microangiopathy with calcifications and cysts

NM_000310.3(PPT1):c.134G>A (p.Cys45Tyr)	PPT1	1	40092498	Ceroid lipofuscinosis neuronal 1
NM_000310.3(PPT1):c.451C>T (p.Arg151Ter)	PPT1	1	40089495	Ceroid lipofuscinosis neuronal 1;Neuronal Ceroid-Lipofuscinosis, Recessive;not provided
NM_000310.3(PPT1):c.424C>T (p.Gln142Ter)	PPT1	1	40091338	Ceroid lipofuscinosis neuronal 1;not provided
NM_001909.4(CTSD):c.470C>T (p.Ser157Leu)	CTSD	11	1758970	Ceroid lipofuscinosis neuronal 10
NM_001909.4(CTSD):c.1196G>A (p.Arg399His)	CTSD	11	1753546	Ceroid lipofuscinosis neuronal 10
NM_000391.3(TPP1):c.616C>T (p.Arg206Cys)	TPP1	11	6617046	Ceroid lipofuscinosis neuronal 2
NM_000391.3(TPP1):c.640C>T (p.Gln214Ter)	TPP1	11	6617022	Ceroid lipofuscinosis neuronal 2
NM_000391.3(TPP1):c.622C>T (p.Arg208Ter)	TPP1	11	6617040	Ceroid lipofuscinosis neuronal 2;Inborn genetic diseases;Neuronal ceroid lipofuscinosis;not provided
NM_000391.3(TPP1):c.1094G>A (p.Cys365Tyr)	TPP1	11	6616056	Ceroid lipofuscinosis neuronal 2;not provided
NM_000391.3(TPP1):c.1340G>A (p.Arg447His)	TPP1	11	6615256	Ceroid lipofuscinosis neuronal 2;not provided
NM_000391.3(TPP1):c.1379G>A (p.Trp460Ter)	TPP1	11	6615217	Ceroid lipofuscinosis neuronal 2;not provided
NM_006493.2(CLN5):c.225G>A (p.Trp75Ter)	CLN5	13	76992176	Ceroid lipofuscinosis neuronal 5
NM_006493.2(CLN5):c.835G>A (p.Asp279Asn)	CLN5	13	77000580	Ceroid lipofuscinosis neuronal 5
NM_006493.2(CLN5):c.335G>A (p.Arg112His)	CLN5	13	76995077	Ceroid lipofuscinosis neuronal 5
NM_006493.2(CLN5):c.377G>A (p.Cys126Tyr)	CLN5	13	76995119	Ceroid lipofuscinosis neuronal 5
NM_006493.2(CLN5):c.935G>A (p.Ser312Asn)	CLN5	13	77000680	Ceroid lipofuscinosis neuronal 5
NM_006493.2(CLN5):c.694C>T (p.Gln232Ter)	CLN5	13	76996109	Ceroid lipofuscinosis neuronal 5
NM_017882.2(CLN6):c.368G>A (p.Gly123Asp)	CLN6	15	68211793	Ceroid lipofuscinosis neuronal 6
NM_000543.4(SMPD1):c.739G>A (p.Gly247Ser)	SMPD1	11	6391804	Ceroid lipofuscinosis neuronal 6;Niemann-Pick disease, type A
NM_152778.2(MFSD8):c.929G>A (p.Gly310Asp)	MFSD8	4	127930752	Ceroid lipofuscinosis neuronal 7
NM_152778.2(MFSD8):c.1286G>A (p.Gly429Asp)	MFSD8	4	127921588	Ceroid lipofuscinosis neuronal 7
NM_152778.2(MFSD8):c.1235C>T (p.Pro412Leu)	MFSD8	4	127921639	Ceroid lipofuscinosis neuronal 7
NM_152778.2(MFSD8):c.1444C>T (p.Arg482Ter)	MFSD8	4	127920743	Ceroid lipofuscinosis neuronal 7;not provided
NM_018941.3(CLN8):c.610C>T (p.Arg204Cys)	CLN8	8	1780316	Ceroid lipofuscinosis neuronal 8
NM_003793.3(CTSF):c.1439C>T (p.Ser480Leu)	CTSF	11	66563949	Ceroid lipofuscinosis, neuronal, 13
NM_000086.2(CLN3):c.494G>A (p.Gly165Glu)	CLN3	16	28486617	Ceroid lipofuscinosis, neuronal, 3, protracted
NM_000086.2(CLN3):c.883G>A (p.Glu295Lys)	CLN3	16	28482500	Ceroid lipofuscinosis, neuronal, 3, protracted;Juvenile neuronal ceroid lipofuscinosis
NM_000081.3(LYST):c.1540C>T (p.Arg514Ter)	LYST	1	235809278	ChÃ©diak-Higashi syndrome
NM_000081.3(LYST):c.8428G>A (p.Glu2810Lys)	LYST	1	235734590	ChÃ©diak-Higashi syndrome
NM_000081.3(LYST):c.8583G>A (p.Trp2861Ter)	LYST	1	235733859	ChÃ©diak-Higashi syndrome
NM_000081.3(LYST):c.11173G>A (p.Gly3725Arg)	LYST	1	235664487	ChÃ©diak-Higashi syndrome
NM_000081.3(LYST):c.5506C>T (p.Arg1836Ter)	LYST	1	235775041	ChÃ©diak-Higashi syndrome
NM_000081.3(LYST):c.3622C>T (p.Gln1208Ter)	LYST	1	235802998	ChÃ©diak-Higashi syndrome
NM_000081.3(LYST):c.925C>T (p.Arg309Ter)	LYST	1	235809893	ChÃ©diak-Higashi syndrome
NM_003221.3(TFAP2B):c.898C>T (p.Arg300Cys)	TFAP2B	6	50838051	Char syndrome
NM_003221.3(TFAP2B):c.706C>T (p.Arg236Cys)	TFAP2B	6	50836165	Char syndrome
NM_003221.3(TFAP2B):c.854G>A (p.Arg285Gln)	TFAP2B	6	50838007	Char syndrome
NM_003221.3(TFAP2B):c.601+5G>A	TFAP2B	6	50828684	Char syndrome;Patent ductus arteriosus 2
NM_138361.5(LRSAM1):c.1913-1G>A	LRSAM1	9	127501009	Charcot-Marie-Tooth disease
NM_021625.4(TRPV4):c.557G>A (p.Arg186Gln)	TRPV4	12	109808298	Charcot-Marie-Tooth disease type 2C;Distal spinal muscular atrophy, congenital nonprogressive;Neuromuscular Diseases;not provided
NM_021625.4(TRPV4):c.694C>T (p.Arg232Cys)	TRPV4	12	109803009	Charcot-Marie-Tooth disease type 2C;Distal spinal muscular atrophy, congenital nonprogressive;Neuromuscular Diseases;Skeletal dysplasia;not provided
NM_021625.4(TRPV4):c.947G>A (p.Arg316His)	TRPV4	12	109798819	Charcot-Marie-Tooth disease type 2C;EMG abnormality;Lower limb amyotrophy;Neuromuscular Diseases;Talipes equinovarus

NM_002047.3(GARS):c.893C>T (p.Pro298Leu)	GARS	7	30612107	Charcot-Marie-Tooth disease type 2D
NM_002047.3(GARS):c.1904C>T (p.Ser635Leu)	GARS	7	30632247	Charcot-Marie-Tooth disease type 2D
NM_002047.3(GARS):c.1660G>A (p.Asp554Asn)	GARS	7	30626280	Charcot-Marie-Tooth disease type 2D;Distal hereditary motor neuronopathy type 5
NM_006158.4(NEFL):c.1261C>T (p.Arg421Ter)	NEFL	8	24953704	Charcot-Marie-Tooth disease type 2E
NM_006158.4(NEFL):c.64C>T (p.Pro22Ser)	NEFL	8	24956452	Charcot-Marie-Tooth disease type 2E;Charcot-Marie-Tooth disease, type 1C;Decreased nerve conduction velocity;Distal lower limb muscle weakness;Distal muscle weakness;Hand muscle atrophy;Peripheral demyelination;Peripheral neuropathy;Pes cavus;not provided
NM_001540.3(HSPB1):c.406C>T (p.Arg136Trp)	HSPB1	7	76303843	Charcot-Marie-Tooth disease type 2F
NM_001540.3(HSPB1):c.404C>T (p.Ser135Phe)	HSPB1	7	76303841	Charcot-Marie-Tooth disease type 2F;Distal hereditary motor neuronopathy type 2B
NM_001540.3(HSPB1):c.379C>T (p.Arg127Trp)	HSPB1	7	76303816	Charcot-Marie-Tooth disease type 2F;Distal hereditary motor neuronopathy type 2B
NM_001540.3(HSPB1):c.523C>T (p.Gln175Ter)	HSPB1	7	76304078	Charcot-Marie-Tooth disease type 2F;not provided
NM_000530.7(MPZ):c.131C>T (p.Ser44Phe)	MPZ	1	161307361	Charcot-Marie-Tooth disease type 2I;Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_018972.2(GDAP1):c.719G>A (p.Cys240Tyr)	GDAP1	8	74364009	Charcot-Marie-Tooth disease type 2K
NM_018972.2(GDAP1):c.821C>T (p.Pro274Leu)	GDAP1	8	74364111	Charcot-Marie-Tooth disease type 2K
NM_018972.2(GDAP1):c.715C>T (p.Leu239Phe)	GDAP1	8	74364005	Charcot-Marie-Tooth disease type 2K;Charcot-Marie-Tooth disease, recessive intermediate A;Charcot-Marie-Tooth disease, type 4A;Elevated alkaline phosphatase;Elevated serum creatine phosphokinase;Peripheral axonal neuropathy;Polyneuropathy;Sensory neuropathy;not provided
NM_018972.2(GDAP1):c.692C>T (p.Pro231Leu)	GDAP1	8	74363051	Charcot-Marie-Tooth disease type 2K;not provided
NM_138361.5(LRSAM1):c.1914G>A (p.Glu638=)	LRSAM1	9	127501011	Charcot-Marie-Tooth disease type 2P
NM_138361.5(LRSAM1):c.1279C>T (p.Arg427Ter)	LRSAM1	9	127487695	Charcot-Marie-Tooth disease type 2P
NM_138361.5(LRSAM1):c.2081G>A (p.Cys694Tyr)	LRSAM1	9	127502808	Charcot-Marie-Tooth disease type 2P
NM_030962.3(SBF2):c.2875C>T (p.Gln959Ter)	SBF2	11	9847015	Charcot-Marie-Tooth disease type 4B2 with early-onset glaucoma
NM_030962.3(SBF2):c.3586C>T (p.Arg1196Ter)	SBF2	11	9832290	Charcot-Marie-Tooth disease type 4B2 with early-onset glaucoma
NM_030962.3(SBF2):c.1459C>T (p.Arg487Ter)	SBF2	11	9968482	Charcot-Marie-Tooth disease type 4B2 with early-onset glaucoma
NM_025137.3(SPG11):c.592C>T (p.Gln198Ter)	SPG11	15	44659154	Charcot-Marie-Tooth disease, axonal type 2X
NM_025137.3(SPG11):c.1621C>T (p.Gln541Ter)	SPG11	15	44633619	Charcot-Marie-Tooth disease, axonal type 2X
NM_025137.3(SPG11):c.6100C>T (p.Arg2034Ter)	SPG11	15	44573652	Charcot-Marie-Tooth disease, axonal type 2X;Spastic paraplegia 11, autosomal recessive
NM_014874.3(MFN2):c.2119C>T (p.Arg707Trp)	MFN2	1	12009641	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL RECESSIVE, TYPE 2A2B;Charcot-Marie-Tooth disease, type 2;Charcot-Marie-Tooth disease, type 2A2;not provided
NM_014874.3(MFN2):c.1085C>T (p.Thr362Met)	MFN2	1	12002028	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL RECESSIVE, TYPE 2A2B;Charcot-Marie-Tooth disease, type 2A2
NM_004637.5(RAB7A):c.385C>T (p.Leu129Phe)	RAB7A	3	128806576	Charcot-Marie-Tooth disease, axonal, type 2b;not provided
NM_004637.5(RAB7A):c.484G>A (p.Val162Met)	RAB7A	3	128807627	Charcot-Marie-Tooth disease, axonal, type 2b;not provided
NM_002180.2(IGHMBP2):c.1582G>A (p.Ala528Thr)	IGHMBP2	11	68934508	Charcot-Marie-Tooth disease, axonal, type 2S;not provided
NM_002180.2(IGHMBP2):c.2560C>T (p.Gln854Ter)	IGHMBP2	11	68937040	Charcot-Marie-Tooth disease, axonal, type 2S;Spinal muscular atrophy, distal, autosomal recessive, 1
NM_002180.2(IGHMBP2):c.1478C>T (p.Thr493Ile)	IGHMBP2	11	68933854	Charcot-Marie-Tooth disease, axonal, type 2S;Spinal muscular atrophy, distal, autosomal recessive, 1;not provided
NG_051105.1:g.97864G>A	MME	3	155116987	Charcot-Marie-Tooth disease, axonal, type 2T
NM_000902.3(MME):c.661C>T (p.Gln221Ter)	MME	3	155118752	Charcot-Marie-Tooth disease, axonal, type 2T
NM_000902.3(MME):c.71G>A (p.Trp24Ter)	MME	3	155084238	Charcot-Marie-Tooth disease, axonal, type 2T
NM_002109.5(HARS):c.410G>A (p.Arg137Gln)	HARS	5	140679114	Charcot-Marie-Tooth disease, axonal, type 2w
NM_002109.5(HARS):c.395C>T (p.Thr132Ile)	HARS	5	140679789	Charcot-Marie-Tooth disease, axonal, type 2w
NM_007126.3(VCP):c.553G>A (p.Glu185Lys)	VCP	9	35065274	Charcot-Marie-Tooth disease, axonal, type 2y
NM_007126.3(VCP):c.290G>A (p.Gly97Glu)	VCP	9	35067903	Charcot-Marie-Tooth disease, axonal, type 2y
NM_001303256.2(MORC2):c.754C>T (p.Arg252Trp)	MORC2	22	30941503	Charcot-Marie-Tooth disease, axonal, type 2z

NM_001303256.2(MORC2):c.260C>T (p.Ser87Leu)	MORC2	22	30949809	Charcot-Marie-Tooth disease, axonal, type 2z
NM_018972.2(GDAP1):c.487C>T (p.Gln163Ter)	GDAP1	8	74361886	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, autosomal recessive;Charcot-Marie-Tooth disease, type 4A;Neuropathy, axonal, with vocal cord paresis, autosomal recessive;not provided
NM_000530.7(MPZ):c.188C>T (p.Ser63Phe)	MPZ	1	161307304	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.308G>A (p.Gly103Glu)	MPZ	1	161306848	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.367G>A (p.Gly123Ser)	MPZ	1	161306789	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.241C>T (p.His81Tyr)	MPZ	1	161306915	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.649C>T (p.Pro217Ser)	MPZ	1	161305974	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.499G>A (p.Gly167Arg)	MPZ	1	161306414	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.487G>A (p.Gly163Arg)	MPZ	1	161306426	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.181G>A (p.Asp61Asn)	MPZ	1	161307311	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.410G>A (p.Gly137Asp)	MPZ	1	161306746	Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_000530.7(MPZ):c.233C>T (p.Ser78Leu)	MPZ	1	161307259	Charcot-Marie-Tooth disease, demyelinating, type 1b;Charcot-Marie-Tooth disease, type 1b, with focally folded myelin sheaths;not provided
NM_000530.7(MPZ):c.409G>A (p.Gly137Ser)	MPZ	1	161306747	Charcot-Marie-Tooth disease, demyelinating, type 1b;Charcot-Marie-Tooth disease, type I
NM_000530.7(MPZ):c.293G>A (p.Arg98His)	MPZ	1	161306863	Charcot-Marie-Tooth disease, demyelinating, type 1b;Charcot-Marie-Tooth disease, type I;Decreased nerve conduction velocity;Distal lower limb amyotrophy;Distal muscle weakness;Pes cavus;Sensory neuropathy;not provided
NM_000530.7(MPZ):c.292C>T (p.Arg98Cys)	MPZ	1	161306864	Charcot-Marie-Tooth disease, demyelinating, type 1b;not provided
NM_000399.4(EGR2):c.1225C>T (p.Arg409Trp)	EGR2	10	62813413	Charcot-Marie-Tooth disease, demyelinating, type 1d
NM_000399.4(EGR2):c.1076G>A (p.Arg359Gln)	EGR2	10	62813562	Charcot-Marie-Tooth disease, demyelinating, type 1d
NM_000399.4(EGR2):c.1142G>A (p.Arg381His)	EGR2	10	62813496	Charcot-Marie-Tooth disease, demyelinating, type 1d
NM_000399.4(EGR2):c.1075C>T (p.Arg359Trp)	EGR2	10	62813563	Charcot-Marie-Tooth disease, demyelinating, type 1d;Charcot-Marie-Tooth disease, type I;Dejerine-Sottas disease;Dejerine-sottas neuropathy, autosomal dominant
NM_006158.4(NEFL):c.446C>T (p.Ala149Val)	NEFL	8	24956070	Charcot-Marie-Tooth disease, demyelinating, type 1f;not provided
NM_181882.2(PRX):c.586C>T (p.Arg196Ter)	PRX	19	40397766	Charcot-Marie-Tooth disease, demyelinating, type 4f;Dejerine-Sottas disease
NM_181882.2(PRX):c.3208C>T (p.Arg1070Ter)	PRX	19	40395144	Charcot-Marie-Tooth disease, demyelinating, type 4f;Dejerine-Sottas disease
NM_181882.2(PRX):c.1951G>A (p.Asp651Asn)	PRX	19	40396401	Charcot-Marie-Tooth disease, demyelinating, type 4f;not provided
NM_003680.3(YARS):c.121G>A (p.Gly41Arg)	YARS	1	32810994	Charcot-Marie-Tooth disease, dominant intermediate C
NM_003680.3(YARS):c.586G>A (p.Glu196Lys)	YARS	1	32797768	Charcot-Marie-Tooth disease, dominant intermediate C
NM_021629.3(GNB4):c.158G>A (p.Gly53Asp)	GNB4	3	179419444	Charcot-Marie-Tooth disease, dominant intermediate F
NM_018972.2(GDAP1):c.980G>A (p.Gly327Asp)	GDAP1	8	74364270	Charcot-Marie-Tooth disease, recessive intermediate A
NM_018972.2(GDAP1):c.844C>T (p.Arg282Cys)	GDAP1	8	74364134	Charcot-Marie-Tooth disease, recessive intermediate A;not provided
NM_020631.4(PLEKHG5):c.1988C>T (p.Thr663Met)	PLEKHG5	1	6469396	Charcot-Marie-Tooth disease, recessive intermediate c
NM_001136472.1(LITAF):c.430G>A (p.Val144Met)	LITAF	16	11549693	Charcot-Marie-Tooth disease, type 1C
NM_004862.3(LITAF):c.403C>T (p.Pro135Ser)	LITAF	16	11549720	Charcot-Marie-Tooth disease, type 1C
NM_004862.3(LITAF):c.385G>A (p.Ala129Thr)	LITAF	16	11549738	Charcot-Marie-Tooth disease, type 1C
NM_004862.3(LITAF):c.334G>A (p.Gly112Ser)	LITAF	16	11553576	Charcot-Marie-Tooth disease, type 1C;not provided
NM_170707.3(LMNA):c.928C>T (p.Gln310Ter)	LMNA	1	156135304	Charcot-Marie-Tooth disease, type 2
NM_014874.3(MFN2):c.703C>T (p.Gln235Ter)	MFN2	1	11998873	Charcot-Marie-Tooth disease, type 2
NM_014874.3(MFN2):c.314C>T (p.Thr105Met)	MFN2	1	11996158	Charcot-Marie-Tooth disease, type 2;Charcot-Marie-Tooth disease, type 2A2
NM_014874.3(MFN2):c.707C>T (p.Thr236Met)	MFN2	1	11998877	Charcot-Marie-Tooth disease, type 2;Charcot-Marie-Tooth disease, type 2A2;Distal lower limb amyotrophy;Distal muscle weakness;Peripheral axonal neuropathy

NM_014874.3(MFN2):c.1090C>T (p.Arg364Trp)	MFN2	1	12002033	Charcot-Marie-Tooth disease, type 2;Charcot-Marie-Tooth disease, type 2A2;Hereditary motor and sensory neuropathy with optic atrophy;not provided
NM_014874.3(MFN2):c.281G>A (p.Arg94Gln)	MFN2	1	11992660	Charcot-Marie-Tooth disease, type 2;Charcot-Marie-Tooth disease, type 2A2;not provided
NM_005572.3(LMNA):c.673C>T (p.Arg225Ter)	LMNA	1	156134838	Charcot-Marie-Tooth disease, type 2;Dilated cardiomyopathy 1A;Limb-girdle muscular dystrophy, type 1B;Primary dilated cardiomyopathy;not provided
NM_170707.3(LMNA):c.1445G>A (p.Arg482Gln)	LMNA	1	156136985	Charcot-Marie-Tooth disease, type 2;Emery-Dreifuss muscular dystrophy 3, autosomal recessive;Familial partial lipodystrophy 2;Partial lipodystrophy, Dunnigan;Primary dilated cardiomyopathy;not provided
NM_170707.3(LMNA):c.1146C>T (p.Gly382=)	LMNA	1	156136110	Charcot-Marie-Tooth disease, type 2;Limb-girdle muscular dystrophy;not provided
NM_170707.3(LMNA):c.1063C>T (p.Gln355Ter)	LMNA	1	156136027	Charcot-Marie-Tooth disease, type 2;not provided
NM_014874.3(MFN2):c.1091G>A (p.Arg364Gln)	MFN2	1	12002034	Charcot-Marie-Tooth disease, type 2;not provided
NM_170707.3(LMNA):c.1129C>T (p.Arg377Cys)	LMNA	1	156136093	Charcot-Marie-Tooth disease, type 2;Primary dilated cardiomyopathy;not provided
NM_170707.3(LMNA):c.1294C>T (p.Gln432Ter)	LMNA	1	156136350	Charcot-Marie-Tooth disease, type 2;Primary dilated cardiomyopathy;not provided
NM_170707.3(LMNA):c.244G>A (p.Glu82Lys)	LMNA	1	156115162	Charcot-Marie-Tooth disease, type 2;Primary dilated cardiomyopathy;not provided
NM_001127660.1(MFN2):c.746C>T (p.Ser249Phe)	MFN2	1	11999025	Charcot-Marie-Tooth disease, type 2A2
NM_001127660.1(MFN2):c.730G>A (p.Val244Met)	MFN2	1	11999009	Charcot-Marie-Tooth disease, type 2A2
NM_014874.3(MFN2):c.839G>A (p.Arg280His)	MFN2	1	12001423	Charcot-Marie-Tooth disease, type 2A2;not provided
NM_001127660.1(MFN2):c.775C>T (p.Arg259Cys)	MFN2	1	11999054	Charcot-Marie-Tooth disease, type 2A2;not provided
NM_001190716.1(DNM2):c.1072G>A (p.Gly358Arg)	DNM2	19	10793799	Charcot-Marie-Tooth disease, type 2M;DNM2-related intermediate Charcot-Marie-Tooth neuropathy;not provided
NM_018972.2(GDAP1):c.92G>A (p.Trp31Ter)	GDAP1	8	74350553	Charcot-Marie-Tooth disease, type 4A
NM_018972.2(GDAP1):c.482G>A (p.Arg161His)	GDAP1	8	74360308	Charcot-Marie-Tooth disease, type 4A
NM_018972.2(GDAP1):c.373C>T (p.Arg125Ter)	GDAP1	8	74360199	Charcot-Marie-Tooth disease, type 4A
NM_018972.2(GDAP1):c.579+1G>A	GDAP1	8	74361979	Charcot-Marie-Tooth disease, type 4A
NM_016156.5(MTMR2):c.1276C>T (p.Gln426Ter)	MTMR2	11	95845063	Charcot-Marie-Tooth disease, type 4B1
NM_016156.5(MTMR2):c.1444C>T (p.Gln482Ter)	MTMR2	11	95841652	Charcot-Marie-Tooth disease, type 4B1
NM_024577.3(SH3TC2):c.1586G>A (p.Arg529His)	SH3TC2	5	149028146	Charcot-Marie-Tooth disease, type 4C
NM_024577.3(SH3TC2):c.1178-1G>A	SH3TC2	5	149028555	Charcot-Marie-Tooth disease, type 4C
NM_024577.3(SH3TC2):c.3601C>T (p.Gln1201Ter)	SH3TC2	5	149006955	Charcot-Marie-Tooth disease, type 4C
NM_024577.3(SH3TC2):c.920G>A (p.Trp307Ter)	SH3TC2	5	149038376	Charcot-Marie-Tooth disease, type 4C
NM_024577.3(SH3TC2):c.1969G>A (p.Glu657Lys)	SH3TC2	5	149027763	Charcot-Marie-Tooth disease, type 4C;Charcot-Marie-Tooth disease, type IV
NM_024577.3(SH3TC2):c.2710C>T (p.Arg904Ter)	SH3TC2	5	149027022	Charcot-Marie-Tooth disease, type 4C;Charcot-Marie-Tooth disease, type IV
NM_024577.3(SH3TC2):c.3325C>T (p.Arg1109Ter)	SH3TC2	5	149010272	Charcot-Marie-Tooth disease, type 4C;not provided
NM_024577.3(SH3TC2):c.1972C>T (p.Arg658Cys)	SH3TC2	5	149027760	Charcot-Marie-Tooth disease, type 4C;not provided
NM_001135242.1(NDRG1):c.442C>T (p.Arg148Ter)	NDRG1	8	133258374	Charcot-Marie-Tooth disease, type 4D;Charcot-Marie-Tooth disease, type IV
NM_139241.3(FGD4):c.670C>T (p.Arg224Ter)	FGD4	12	32598566	Charcot-Marie-Tooth disease, type 4H
NM_139241.3(FGD4):c.823C>T (p.Arg275Ter)	FGD4	12	32601410	Charcot-Marie-Tooth disease, type 4H
NM_139241.3(FGD4):c.1325G>A (p.Arg442His)	FGD4	12	32611270	Charcot-Marie-Tooth disease, type 4H
NM_014845.5(FIG4):c.904G>A (p.Glu302Lys)	FIG4	6	109743137	Charcot-Marie-Tooth disease, type 4J
NM_014845.5(FIG4):c.1141C>T (p.Arg381Ter)	FIG4	6	109760253	Charcot-Marie-Tooth disease, type 4J
NM_003172.3(SURF1):c.574C>T (p.Arg192Trp)	SURF1	9	133352708	Charcot-Marie-Tooth disease, type 4k;not provided
NM_000304.3(PMP22):c.215C>T (p.Ser72Leu)	PMP22	17	15239575	Charcot-Marie-Tooth disease, type I;Dejerine-Sottas disease;Dejerine-Sottas syndrome, autosomal dominant
NM_000399.4(EGR2):c.1226G>A (p.Arg409Gln)	EGR2	10	62813412	Charcot-Marie-Tooth disease, type I;not provided
NM_000304.3(PMP22):c.65C>T (p.Ser22Phe)	PMP22	17	15260663	Charcot-Marie-Tooth disease, type IA;Hereditary liability to pressure palsies

NM_024577.3(SH3TC2):c.279G>A (p.Lys93=)	SH3TC2	5	149047862	Charcot-Marie-Tooth disease, type IV
NM_024577.3(SH3TC2):c.211C>T (p.Gln71Ter)	SH3TC2	5	149047930	Charcot-Marie-Tooth disease, type IV
NM_024577.3(SH3TC2):c.3154C>T (p.Arg1052Ter)	SH3TC2	5	149012634	Charcot-Marie-Tooth disease, type IV
NM_016156.5(MTMR2):c.832C>T (p.Gln278Ter)	MTMR2	11	95849835	Charcot-Marie-Tooth disease, type IV
NM_030962.3(SBF2):c.2536+1G>A	SBF2	11	9853539	Charcot-Marie-Tooth disease, type IV
NM_139241.3(FGD4):c.1729C>T (p.Arg577Ter)	FGD4	12	32625747	Charcot-Marie-Tooth disease, type IV
NM_005391.4(PDK3):c.473G>A (p.Arg158His)	PDK3	X	24503479	Charcot-Marie-Tooth disease, X-linked dominant, 6
NM_170707.3(LMNA):c.892C>T (p.Arg298Cys)	LMNA	1	156135268	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease type 2B1;not provided
NM_021625.4(TRPV4):c.806G>A (p.Arg269His)	TRPV4	12	109800665	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease type 2C;Distal spinal muscular atrophy, congenital nonprogressive;Neuromuscular Diseases;not provided
NM_021625.4(TRPV4):c.805C>T (p.Arg269Cys)	TRPV4	12	109800666	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease type 2C;Distal spinal muscular atrophy, congenital nonprogressive;Neuromuscular Diseases;Scapuloperoneal spinal muscular atrophy
NM_021625.4(TRPV4):c.943C>T (p.Arg315Trp)	TRPV4	12	109798823	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease type 2C;Distal spinal muscular atrophy, congenital nonprogressive;Neuromuscular Diseases;Scapuloperoneal spinal muscular atrophy;not provided
NM_021625.4(TRPV4):c.946C>T (p.Arg316Cys)	TRPV4	12	109798820	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease type 2C;Neuromuscular Diseases;Scapuloperoneal spinal muscular atrophy;not provided
NM_000530.7(MPZ):c.371C>T (p.Thr124Met)	MPZ	1	161306785	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease type 2J;Charcot-Marie-Tooth disease, demyelinating, type 1b
NM_018972.2(GDAP1):c.358C>T (p.Arg120Trp)	GDAP1	8	74360184	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease type 2K;Charcot-Marie-Tooth disease, type 4A;not provided
NM_001605.2(AARS):c.986G>A (p.Arg329His)	AARS	16	70268356	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease, type 2;Charcot-Marie-Tooth disease, type 2N
NM_014874.3(MFN2):c.310C>T (p.Arg104Trp)	MFN2	1	11992689	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease, type 2A2;Hereditary motor and sensory neuropathy with optic atrophy;not provided
NM_002972.3(SBF1):c.1327G>A (p.Asp443Asn)	SBF1	22	50465006	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease, type 4B3
NM_024577.3(SH3TC2):c.2860C>T (p.Arg954Ter)	SH3TC2	5	149026872	Charcot-Marie-Tooth disease;Charcot-Marie-Tooth disease, type 4C;Charcot-Marie-Tooth disease, type IV;Mononeuropathy of the median nerve, mild;SH3TC2-Related Disorders;not provided
NM_170707.3(LMNA):c.1579C>T (p.Arg527Cys)	LMNA	1	156137203	Charcot-Marie-Tooth disease;Hutchinson-Gilford syndrome;Mandibuloacral dysostosis;not provided
NM_170707.3(LMNA):c.1411C>T (p.Arg471Cys)	LMNA	1	156136951	Charcot-Marie-Tooth disease;Mandibuloacral dysplasia with type A lipodystrophy, atypical;not provided
NM_170707.3(LMNA):c.1908C>T (p.Ser636=)	LMNA	1	156138697	Charcot-Marie-Tooth disease;not provided
NM_001039550.1(DNAJB2):c.229+1G>A	DNAJB2	2	219281772	Charcot-Marie-Tooth disease;Spinal muscular atrophy, distal, autosomal recessive, 5
NM_002180.2(IGHMBP2):c.1738G>A (p.Val580Ile)	IGHMBP2	11	68935404	Charcot-Marie-Tooth disease;Werdnig-Hoffmann disease
NM_000166.5(GJB1):c.116C>T (p.Ala39Val)	GJB1	X	71223823	Charcot-Marie-Tooth Neuropathy X
NM_000166.5(GJB1):c.425G>A (p.Arg142Gln)	GJB1	X	71224132	Charcot-Marie-Tooth Neuropathy X
NM_000166.5(GJB1):c.9G>A (p.Trp3Ter)	GJB1	X	71223716	Charcot-Marie-Tooth Neuropathy X
NM_000166.5(GJB1):c.65G>A (p.Arg22Gln)	GJB1	X	71223772	Charcot-Marie-Tooth Neuropathy X
NM_000166.5(GJB1):c.547C>T (p.Arg183Cys)	GJB1	X	71224254	Charcot-Marie-Tooth Neuropathy X;not provided
NM_000166.5(GJB1):c.-103C>T	GJB1	X	71223249	Charcot-Marie-Tooth Neuropathy X;X-linked hereditary motor and sensory neuropathy
NM_000166.5(GJB1):c.424C>T (p.Arg142Trp)	GJB1	X	71224131	Charcot-Marie-Tooth Neuropathy X;X-linked hereditary motor and sensory neuropathy;not provided
NM_000166.5(GJB1):c.658C>T (p.Arg220Ter)	GJB1	X	71224365	Charcot-Marie-Tooth Neuropathy X;X-linked hereditary motor and sensory neuropathy;not provided
NM_000166.5(GJB1):c.283G>A (p.Val95Met)	GJB1	X	71223990	Charcot-Marie-Tooth Neuropathy X;X-linked hereditary motor and sensory neuropathy;not provided
NM_000166.5(GJB1):c.43C>T (p.Arg15Trp)	GJB1	X	71223750	Charcot-Marie-Tooth Neuropathy X;X-linked hereditary motor and sensory neuropathy;not provided
NM_000166.5(GJB1):c.77C>T (p.Ser26Leu)	GJB1	X	71223784	Charcot-Marie-Tooth Neuropathy X;X-linked hereditary motor and sensory neuropathy;not provided

NM_000166.5(GJB1):c.44G>A (p.Arg15Gln)	GJB1	X	71223751	Charcot-Marie-Tooth Neuropathy X;X-linked hereditary motor and sensory neuropathy;not provided
NM_017780.3(CHD7):c.6322G>A (p.Gly2108Arg)	CHD7	8	60853047	CHARGE association
NM_012431.2(SEMA3E):c.2108C>T (p.Ser703Leu)	SEMA3E	7	83367806	CHARGE association
NM_017780.3(CHD7):c.2613+1G>A	CHD7	8	60816502	CHARGE association
NM_017780.3(CHD7):c.4318C>T (p.Gln1440Ter)	CHD7	8	60837800	CHARGE association
NM_017780.3(CHD7):c.4480C>T (p.Arg1494Ter)	CHD7	8	60838202	CHARGE association
NM_017780.3(CHD7):c.6850C>T (p.Arg2284Ter)	CHD7	8	60854437	CHARGE association
NM_017780.3(CHD7):c.7252C>T (p.Arg2418Ter)	CHD7	8	60856532	CHARGE association
NM_017780.3(CHD7):c.7957C>T (p.Arg2653Ter)	CHD7	8	60862322	CHARGE association
NM_017780.3(CHD7):c.8055G>A (p.Trp2685Ter)	CHD7	8	60862631	CHARGE association
NM_017780.3(CHD7):c.2933G>A (p.Trp978Ter)	CHD7	8	60822121	CHARGE association
NM_017780.3(CHD7):c.6995G>A (p.Trp2332Ter)	CHD7	8	60856033	CHARGE association
NM_017780.3(CHD7):c.5023C>T (p.Gln1675Ter)	CHD7	8	60845036	CHARGE association
NM_017780.3(CHD7):c.6292C>T (p.Arg2098Ter)	CHD7	8	60853017	CHARGE association
NM_017780.3(CHD7):c.6955C>T (p.Arg2319Cys)	CHD7	8	60855993	CHARGE association
NM_017780.3(CHD7):c.496C>T (p.Gln166Ter)	CHD7	8	60741928	CHARGE association
NM_017780.3(CHD7):c.3205C>T (p.Arg1069Ter)	CHD7	8	60823843	CHARGE association
NM_017780.3(CHD7):c.3526C>T (p.Gln1176Ter)	CHD7	8	60830325	CHARGE association
NM_017780.3(CHD7):c.6148C>T (p.Arg2050Ter)	CHD7	8	60852873	CHARGE association
NM_017780.3(CHD7):c.1552C>T (p.Gln518Ter)	CHD7	8	60742984	CHARGE association
NM_017780.3(CHD7):c.7312C>T (p.Gln2438Ter)	CHD7	8	60856592	CHARGE association
NM_017780.3(CHD7):c.2501C>T (p.Ser834Phe)	CHD7	8	60816389	CHARGE association;Hypogonadotropic hypogonadism 5 without anosmia
NM_017780.3(CHD7):c.4795C>T (p.Gln1599Ter)	CHD7	8	60841997	CHARGE association;not provided
NM_017780.3(CHD7):c.2839C>T (p.Arg947Ter)	CHD7	8	60822027	CHARGE association;not provided
NM_017780.3(CHD7):c.5405-7G>A	CHD7	8	60850486	CHARGE association;not provided
NM_017780.3(CHD7):c.1480C>T (p.Arg494Ter)	CHD7	8	60742912	CHARGE association;not provided
NM_017780.3(CHD7):c.5458C>T (p.Arg1820Ter)	CHD7	8	60850546	CHARGE association;not provided
NM_017780.3(CHD7):c.6157C>T (p.Arg2053Ter)	CHD7	8	60852882	CHARGE association;not provided
NM_017780.3(CHD7):c.7891C>T (p.Arg2631Ter)	CHD7	8	60862256	CHARGE association;not provided
NM_017780.3(CHD7):c.1090C>T (p.Gln364Ter)	CHD7	8	60742522	CHARGE association;not provided
NM_017780.3(CHD7):c.1369C>T (p.Gln457Ter)	CHD7	8	60742801	CHARGE association;not provided
NM_017780.3(CHD7):c.469C>T (p.Arg157Ter)	CHD7	8	60741901	CHARGE association;not provided
NM_017780.3(CHD7):c.1465C>T (p.Gln489Ter)	CHD7	8	60742897	CHARGE association;not provided
NM_017780.3(CHD7):c.2959C>T (p.Arg987Ter)	CHD7	8	60822504	CHARGE association;not provided
NM_017780.3(CHD7):c.4393C>T (p.Arg1465Ter)	CHD7	8	60838115	CHARGE association;not provided
NM_017780.3(CHD7):c.5833C>T (p.Arg1945Ter)	CHD7	8	60852186	CHARGE association;not provided
NM_017780.3(CHD7):c.6079C>T (p.Arg2027Ter)	CHD7	8	60852682	CHARGE association;not provided
NM_000081.3(LYST):c.4688G>A (p.Arg1563His)	LYST	1	235788701	Chediak-Higashi syndrome, adult type;ChÃ©diak-Higashi syndrome
NM_000081.3(LYST):c.3310C>T (p.Arg1104Ter)	LYST	1	235805826	Chediak-Higashi syndrome, adult type;ChÃ©diak-Higashi syndrome;not provided
NM_000081.3(LYST):c.148C>T (p.Arg50Ter)	LYST	1	235830270	Chediak-Higashi syndrome, childhood type;ChÃ©diak-Higashi syndrome
NM_000081.3(LYST):c.3085C>T (p.Gln1029Ter)	LYST	1	235806051	Chediak-Higashi syndrome, childhood type;ChÃ©diak-Higashi syndrome
NM_007194.3(CHEK2):c.433C>T (p.Arg145Trp)	CHEK2	22	28725254	CHEK2-Related Cancer Susceptibility;Familial cancer of breast;Hereditary cancer-predisposing syndrome;Li Fraumeni syndrome 2;not specified
NM_001129765.1(NSDHL):c.314C>T (p.Ala105Val)	NSDHL	X	152858816	Child syndrome
NM_001129765.1(NSDHL):c.613G>A (p.Gly205Ser)	NSDHL	X	152865888	Child syndrome

NM_001129765.1(NSDHL):c.628C>T (p.Gln210Ter)	NSDHL	X	152865903	Child syndrome	
NM_001129765.1(NSDHL):c.262C>T (p.Arg88Ter)	NSDHL	X	152850418	Child syndrome	
NM_001129765.1(NSDHL):c.370G>A (p.Gly124Ser)	NSDHL	X	152858872	Child syndrome	
NM_015922.2(NSDHL):c.757C>T (p.Gln253Ter)	NSDHL	X	152867641	Child syndrome	
NM_001127500.2(MET):c.3572C>T (p.Thr1191Ile)	MET	7	116778953	Childhood hepatocellular carcinoma	
NM_001127500.2(MET):c.3804G>A (p.Met1268Ile)	MET	7	116783421	Childhood hepatocellular carcinoma;Neoplasm	
NM_000478.5(ALPL):c.407G>A (p.Arg136His)	ALPL	1	21563219	Childhood hypophosphatasia;Hypophosphatasia;Infantile hypophosphatasia	
NM_000478.5(ALPL):c.526G>A (p.Ala176Thr)	ALPL	1	21564094	Childhood hypophosphatasia;Infantile hypophosphatasia	
NM_000478.5(ALPL):c.814C>T (p.Arg272Cys)	ALPL	1	21570326	Childhood hypophosphatasia;Infantile hypophosphatasia	
NM_000784.3(CYP27A1):c.1435C>T (p.Arg479Cys)	CYP27A1	2	218814716	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.845-1G>A	CYP27A1	2	218812923	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.844+1G>A	CYP27A1	2	218812750	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1421G>A (p.Arg474Gln)	CYP27A1	2	218814702	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1420C>T (p.Arg474Trp)	CYP27A1	2	218814701	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1214G>A (p.Arg405Gln)	CYP27A1	2	218814409	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1263+1G>A	CYP27A1	2	218814459	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.434G>A (p.Gly145Glu)	CYP27A1	2	218809755	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1016C>T (p.Thr339Met)	CYP27A1	2	218813095	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1184G>A (p.Arg395His)	CYP27A1	2	218814187	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1213C>T (p.Arg405Trp)	CYP27A1	2	218814408	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1264-1G>A	CYP27A1	2	218814544	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1402C>T (p.Pro468Ser)	CYP27A1	2	218814683	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.379C>T (p.Arg127Trp)	CYP27A1	2	218809700	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.380G>A (p.Arg127Gln)	CYP27A1	2	218809701	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.409C>T (p.Arg137Trp)	CYP27A1	2	218809730	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.433G>A (p.Gly145Arg)	CYP27A1	2	218809754	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.446+1G>A	CYP27A1	2	218809768	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.691C>T (p.Arg231Ter)	CYP27A1	2	218812596	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.745C>T (p.Gln249Ter)	CYP27A1	2	218812650	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.779G>A (p.Trp260Ter)	CYP27A1	2	218812684	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.808C>T (p.Arg270Ter)	CYP27A1	2	218812713	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.410G>A (p.Arg137Gln)	CYP27A1	2	218809731	Cholestanol storage disease	
NM_000784.3(CYP27A1):c.1183C>T (p.Arg395Cys)	CYP27A1	2	218814186	Cholestanol storage disease;not provided	
NM_000784.3(CYP27A1):c.1184+1G>A	CYP27A1	2	218814188	Cholestanol storage disease;not provided	
NM_000784.3(CYP27A1):c.475C>T (p.Gln159Ter)	CYP27A1	2	218812250	Cholestanol storage disease;not provided	
NM_005603.4(ATP8B1):c.2599C>T (p.Arg867Cys)	ATP8B1	18	57661282	Cholestasis of pregnancy	
NM_018849.2(ABCB4):c.430C>T (p.Arg144Ter)	ABCB4	7	87453050	Cholestasis, intrahepatic, of pregnancy 3	
NM_018849.2(ABCB4):c.2869C>T (p.Arg957Ter)	ABCB4	7	87411948	Cholestasis, intrahepatic, of pregnancy 3;Progressive familial intrahepatic cholestasis 3	
NM_005123.3(NR1H4):c.526C>T (p.Arg176Ter)	NR1H4	12	100532538	Cholestasis, progressive familial intrahepatic, 5;Progressive intrahepatic cholestasis	
NM_000349.2(STAR):c.772C>T (p.Gln258Ter)	STAR	8	38144359	Cholesterol monooxygenase (side-chain cleaving) deficiency	
NM_000349.2(STAR):c.749G>A (p.Trp250Ter)	STAR	8	38144382	Cholesterol monooxygenase (side-chain cleaving) deficiency	
NM_000349.2(STAR):c.653C>T (p.Ala218Val)	STAR	8	38145313	Cholesterol monooxygenase (side-chain cleaving) deficiency	
NM_000349.2(STAR):c.545G>A (p.Arg182His)	STAR	8	38146068	Cholesterol monooxygenase (side-chain cleaving) deficiency	
NM_000349.2(STAR):c.559G>A (p.Val187Met)	STAR	8	38146054	Cholesterol monooxygenase (side-chain cleaving) deficiency	
NM_000349.2(STAR):c.562C>T (p.Arg188Cys)	STAR	8	38146051	Cholesterol monooxygenase (side-chain cleaving) deficiency	

NM_000349.2(STAR):c.577C>T (p.Arg193Ter)	STAR	8	38146036	Cholesterol monooxygenase (side-chain cleaving) deficiency
NM_054027.4(ANKH):c.14C>T (p.Pro5Leu)	ANKH	5	14871434	Chondrocalcinosis 2
NM_054027.4(ANKH):c.1165G>A (p.Gly389Arg)	ANKH	5	14713644	Chondrocalcinosis 2;Craniometaphyseal dysplasia, autosomal dominant
NM_001256794.1(BMPR1B):c.657G>A (p.Trp219Ter)	BMPR1B	4	95129933	Chondrodysplasia acromesomelic with genital anomalies
NM_001203.2(BMPR1B):c.91C>T (p.Arg31Cys)	BMPR1B	4	95104515	Chondrodysplasia acromesomelic with genital anomalies
NM_000316.2(PTH1R):c.1148G>A (p.Arg383Gln)	PTH1R	3	46901797	Chondrodysplasia Blomstrand type
NM_000316.2(PTH1R):c.395C>T (p.Pro132Leu)	PTH1R	3	46897936	Chondrodysplasia Blomstrand type
NM_000316.2(PTH1R):c.310C>T (p.Arg104Ter)	PTH1R	3	46895866	Chondrodysplasia Blomstrand type
NM_000047.2(ARSE):c.349G>A (p.Gly117Arg)	ARSE	X	2953224	Chondrodysplasia punctata 1, X-linked recessive
NM_000047.2(ARSE):c.1475G>A (p.Cys492Tyr)	ARSE	X	2935127	Chondrodysplasia punctata 1, X-linked recessive
NM_000047.2(ARSE):c.1732C>T (p.Pro578Ser)	ARSE	X	2934870	Chondrodysplasia punctata 1, X-linked recessive
NM_000047.2(ARSE):c.1442C>T (p.Thr481Met)	ARSE	X	2935160	Chondrodysplasia punctata 1, X-linked recessive
NM_000047.2(ARSE):c.1743G>A (p.Trp581Ter)	ARSE	X	2934859	Chondrodysplasia punctata 1, X-linked recessive;not provided
NM_006579.2(EBP):c.87G>A (p.Trp29Ter)	EBP	X	48523858	Chondrodysplasia punctata 2 X-linked dominant
NM_006579.2(EBP):c.187C>T (p.Arg63Ter)	EBP	X	48523958	Chondrodysplasia punctata 2 X-linked dominant
NM_006579.2(EBP):c.238G>A (p.Glu80Lys)	EBP	X	48524009	Chondrodysplasia punctata 2 X-linked dominant
NM_006579.2(EBP):c.386G>A (p.Trp129Ter)	EBP	X	48527202	Chondrodysplasia punctata 2 X-linked dominant
NM_006579.2(EBP):c.523C>T (p.Gln175Ter)	EBP	X	48528287	Chondrodysplasia punctata 2 X-linked dominant
NM_006579.2(EBP):c.587G>A (p.Trp196Ter)	EBP	X	48528351	Chondrodysplasia punctata 2 X-linked dominant
NM_006579.2(EBP):c.440G>A (p.Arg147His)	EBP	X	48527256	Chondrodysplasia punctata 2 X-linked dominant
NM_006579.2(EBP):c.182G>A (p.Trp61Ter)	EBP	X	48523953	Chondrodysplasia punctata 2 X-linked dominant
NM_006579.2(EBP):c.328C>T (p.Arg110Ter)	EBP	X	48527015	Chondrodysplasia punctata 2 X-linked dominant
NM_017813.4(IMPAD1):c.529G>A (p.Asp177Asn)	IMPAD1	8	56980056	Chondrodysplasia with joint dislocations, GPAPP type
NM_017813.4(IMPAD1):c.559C>T (p.Arg187Ter)	IMPAD1	8	56978137	Chondrodysplasia with joint dislocations, GPAPP type
NM_001166136.1(EVC2):c.955C>T (p.Arg319Ter)	EVC2	4	5640789	Chondroectodermal dysplasia
NM_001166136.1(EVC2):c.1615C>T (p.Gln539Ter)	EVC2	4	5628590	Chondroectodermal dysplasia
NM_001166136.1(EVC2):c.3025C>T (p.Gln1009Ter)	EVC2	4	5576247	Chondroectodermal dysplasia
NM_001166136.1(EVC2):c.2608C>T (p.Arg870Trp)	EVC2	4	5584832	Chondroectodermal dysplasia
NM_153717.2(EVC):c.2635C>T (p.Gln879Ter)	EVC	4	5808274	Chondroectodermal dysplasia
NM_147127.4(EVC2):c.2653C>T (p.Arg885Ter)	EVC2	4	5618531	Chondroectodermal dysplasia
NM_147127.4(EVC2):c.2263C>T (p.Gln755Ter)	EVC2	4	5622775	Chondroectodermal dysplasia
NM_147127.4(EVC2):c.3141G>A (p.Trp1047Ter)	EVC2	4	5576371	Chondroectodermal dysplasia
NM_153717.2(EVC):c.1018C>T (p.Arg340Ter)	EVC	4	5748226	Chondroectodermal dysplasia;not provided
NM_014423.3(AFF4):c.772C>T (p.Arg258Trp)	AFF4	5	132934293	Chops syndrome;not provided
NM_014334.3(FRRS1L):c.961C>T (p.Gln321Ter)	FRRS1L	9	109137529	Chorea;Epileptic encephalopathy, early infantile, 37;Progressive encephalopathy;Seizures
NM_014334.3(FRRS1L):c.845G>A (p.Trp282Ter)	FRRS1L	9	109141360	Chorea;Epileptic encephalopathy, early infantile, 37;Progressive encephalopathy;Seizures
NM_033305.2(VPS13A):c.622C>T (p.Arg208Ter)	VPS13A	9	77213240	Choreoacanthocytosis
NM_000322.4(PRPH2):c.515G>A (p.Arg172Gln)	PRPH2	6	42721820	Choroidal dystrophy, central areolar 2;not provided
NM_000322.4(PRPH2):c.514C>T (p.Arg172Trp)	PRPH2	6	42721821	Choroidal dystrophy, central areolar 2;not provided
NM_000322.4(PRPH2):c.424C>T (p.Arg142Trp)	PRPH2	6	42721911	Choroidal dystrophy, central areolar 2;not provided
NM_000390.3(CHM):c.808C>T (p.Arg270Ter)	CHM	X	85958872	Choroideremia
NM_000390.3(CHM):c.715C>T (p.Arg239Ter)	CHM	X	85958965	Choroideremia
NM_000390.3(CHM):c.877C>T (p.Arg293Ter)	CHM	X	85957918	Choroideremia;not provided
NM_001042537.1(SLC9A6):c.1498C>T (p.Arg500Ter)	SLC9A6	X	136024365	Christianson syndrome
NM_001042537.1(SLC9A6):c.604-1G>A	SLC9A6	X	135998481	Christianson syndrome

NM_000545.6(HNF1A):c.92G>A (p.Gly31Asp)	HNF1A	12	120978860	Chromophobe renal cell carcinoma;Clear cell carcinoma of kidney;Maturity-onset diabetes of the young, type 3
NM_015265.3(SATB2):c.715C>T (p.Arg239Ter)	SATB2	2	199349159	Chromosome 2q32-q33 deletion syndrome;not provided
NM_024757.4(EHMT1):c.3502C>T (p.Arg1168Ter)	EHMT1	9	137818100	Chromosome 9q deletion syndrome
NM_024757.4(EHMT1):c.3218G>A (p.Cys1073Tyr)	EHMT1	9	137814468	Chromosome 9q deletion syndrome
NM_024757.4(EHMT1):c.871C>T (p.Arg291Ter)	EHMT1	9	137743418	Chromosome 9q deletion syndrome
NM_024757.4(EHMT1):c.1810C>T (p.Gln604Ter)	EHMT1	9	137776636	Chromosome 9q deletion syndrome
NM_024757.4(EHMT1):c.1858C>T (p.Arg620Ter)	EHMT1	9	137776684	Chromosome 9q deletion syndrome
NM_024757.4(EHMT1):c.2868-1G>A	EHMT1	9	137813005	Chromosome 9q deletion syndrome
NM_024757.4(EHMT1):c.3229C>T (p.Gln1077Ter)	EHMT1	9	137814479	Chromosome 9q deletion syndrome
NM_024757.4(EHMT1):c.3589C>T (p.Arg1197Trp)	EHMT1	9	137834397	Chromosome 9q deletion syndrome
NM_024757.4(EHMT1):c.673C>T (p.Arg225Ter)	EHMT1	9	137728379	Chromosome 9q deletion syndrome;not provided
NM_005745.7(BCAP31):c.97C>T (p.Gln33Ter)	BCAP31	X	153720968	Chromosome Xq28 deletion syndrome
NM_001127221.1(CACNA1A):c.4991G>A (p.Arg1664Gln)	CACNA1A	19	13235693	Chronic and progressive ataxia;Enlarged cisterna magna;Global developmental delay;Spinocerebellar ataxia 6;not provided
NM_000265.5(NCF1):c.125G>A (p.Arg42Gln)	NCF1	7	74777319	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 1
NM_000265.5(NCF1):c.271C>T (p.Gln91Ter)	NCF1	7	74779298	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 1
NM_000265.5(NCF1):c.574G>A (p.Gly192Ser)	NCF1	7	74783061	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 1
NM_000433.3(NCF2):c.304C>T (p.Arg102Ter)	NCF2	1	183577661	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 2
NM_000433.3(NCF2):c.366+1G>A	NCF2	1	183577598	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 2
NM_000433.3(NCF2):c.298C>T (p.Gln100Ter)	NCF2	1	183577667	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 2
NM_000433.3(NCF2):c.383C>T (p.Ala128Val)	NCF2	1	183574605	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 2
NM_000433.3(NCF2):c.230G>A (p.Arg77Gln)	NCF2	1	183586922	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 2
NM_000433.3(NCF2):c.565C>T (p.Gln189Ter)	NCF2	1	183573229	Chronic granulomatous disease, autosomal recessive cytochrome b-positive, type 2
NM_000397.3(CYBB):c.676C>T (p.Arg226Ter)	CYBB	X	37798956	Chronic granulomatous disease, X-linked
NM_000397.3(CYBB):c.625C>T (p.His209Tyr)	CYBB	X	37796092	Chronic granulomatous disease, X-linked;not provided
NM_000397.3(CYBB):c.217C>T (p.Arg73Ter)	CYBB	X	37783565	Chronic granulomatous disease, X-linked;not provided
NM_000397.3(CYBB):c.252G>A (p.Ala84=)	CYBB	X	37783600	Chronic granulomatous disease, X-linked;not provided
NM_013416.3(NCF4):c.314G>A (p.Arg105Gln)	NCF4	22	36867434	Chronic granulomatous disease;Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III
NM_001243133.1(NLRP3):c.907G>A (p.Asp303Asn)	NLRP3	1	247424356	Chronic infantile neurological, cutaneous and articular syndrome;Familial amyloid nephropathy with urticaria AND deafness;Familial cold urticaria;not provided
NM_001615.3(ACTG2):c.113G>A (p.Arg38His)	ACTG2	2	73901424	Chronic intestinal pseudoobstruction
NM_001615.3(ACTG2):c.770G>A (p.Arg257His)	ACTG2	2	73914836	Chronic intestinal pseudoobstruction;Inborn genetic diseases;Megacystis
NM_001615.3(ACTG2):c.769C>T (p.Arg257Cys)	ACTG2	2	73914835	Chronic intestinal pseudoobstruction;Inborn genetic diseases;Megacystis;Visceral myopathy;not provided
NM_007313.2(ABL1):c.763G>A (p.Glu255Lys)	ABL1	9	130862919	Chronic myeloid leukemia;Leukemia, Philadelphia chromosome-positive, resistant to imatinib
NM_007313.2(ABL1):c.1001C>T (p.Thr334Ile)	ABL1	9	130872896	Chronic myeloid leukemia;Leukemia, Philadelphia chromosome-positive, resistant to imatinib;Lymphoblastic leukemia, acute, with lymphomatous features
NM_015559.2(SETBP1):c.2608G>A (p.Gly870Ser)	SETBP1	18	44951948	Chronic myeloid leukemia;Schinzel-Giedion syndrome
NM_013296.4(GPSM2):c.1684C>T (p.Gln562Ter)	GPSM2	1	108924083	Chudley-McCullough syndrome
NM_001033503.2(SAR1B):c.109G>A (p.Gly37Arg)	SAR1B	5	134621002	Chylomicron retention disease
NM_001033503.2(SAR1B):c.409G>A (p.Asp137Asn)	SAR1B	5	134608443	Chylomicron retention disease
NM_001010892.2(RSPH4A):c.1468C>T (p.Arg490Ter)	RSPH4A	6	116628175	Ciliary dyskinesia, primary, 11
NM_001010892.2(RSPH4A):c.460C>T (p.Gln154Ter)	RSPH4A	6	116617083	Ciliary dyskinesia, primary, 11;Primary ciliary dyskinesia

NM_001010892.2(RSPH4A):c.325C>T (p.Gln109Ter)	RSPH4A	6	116616948	Ciliary dyskinesia, primary, 11;Primary ciliary dyskinesia
NM_152732.4(RSPH9):c.52C>T (p.Gln18Ter)	RSPH9	6	43645150	Ciliary dyskinesia, primary, 12
NM_178452.5(DNAAF1):c.811C>T (p.Arg271Ter)	DNAAF1	16	84159744	Ciliary dyskinesia, primary, 13
NM_017950.3(CCDC40):c.1315C>T (p.Gln439Ter)	CCDC40	17	80058649	Ciliary dyskinesia, primary, 15
NM_017950.3(CCDC40):c.1951C>T (p.Gln651Ter)	CCDC40	17	80082020	Ciliary dyskinesia, primary, 15
NM_017950.3(CCDC40):c.1345C>T (p.Arg449Ter)	CCDC40	17	80058885	Ciliary dyskinesia, primary, 15
NM_012472.5(LRRC6):c.574C>T (p.Gln192Ter)	LRRC6	8	132632819	Ciliary dyskinesia, primary, 19
NM_012472.5(LRRC6):c.562C>T (p.Gln188Ter)	LRRC6	8	132632831	Ciliary dyskinesia, primary, 19
NM_001256714.1(DNAAF3):c.469C>T (p.Arg157Ter)	DNAAF3	19	55165427	Ciliary dyskinesia, primary, 2;Kartagener syndrome
NM_144577.3(CCDC114):c.486+1G>A	CCDC114	19	48311552	Ciliary dyskinesia, primary, 20
NM_144577.3(CCDC114):c.1391+5G>A	CCDC114	19	48297995	Ciliary dyskinesia, primary, 20
NM_144577.3(CCDC114):c.742G>A (p.Ala248Thr)	CCDC114	19	48303953	Ciliary dyskinesia, primary, 20;Kartagener syndrome;Primary ciliary dyskinesia
NM_145038.4(DRC1):c.352C>T (p.Gln118Ter)	DRC1	2	26421396	Ciliary dyskinesia, primary, 21;Kartagener syndrome
NM_015896.3(ZMYND10):c.967C>T (p.Gln323Ter)	ZMYND10	3	50342047	Ciliary dyskinesia, primary, 22
NM_021254.3(C21orf59):c.292C>T (p.Arg98Ter)	C21orf59	21	32609853	Ciliary dyskinesia, primary, 26;Kartagener syndrome
NM_021147.4(CCNO):c.961C>T (p.Gln321Ter)	CCNO	5	55231467	Ciliary dyskinesia, primary, 29;Kartagener syndrome
NM_001369.2(DNAH5):c.1828C>T (p.Gln610Ter)	DNAH5	5	13901476	Ciliary dyskinesia, primary, 3
NM_001369.2(DNAH5):c.4348C>T (p.Gln1450Ter)	DNAH5	5	13865675	Ciliary dyskinesia, primary, 3;Kartagener syndrome
NM_031924.5(RSPH3):c.616C>T (p.Gln206Ter)	RSPH3	6	158993853	Ciliary dyskinesia, primary, 32
NM_031924.5(RSPH3):c.1192C>T (p.Arg398Ter)	RSPH3	6	158980867	Ciliary dyskinesia, primary, 32
NM_031924.5(RSPH3):c.1105C>T (p.Arg369Ter)	RSPH3	6	158982502	Ciliary dyskinesia, primary, 32
NM_001286209.1(GAS8):c.925C>T (p.Arg309Ter)	GAS8	16	90037837	Ciliary dyskinesia, primary, 33
NM_001286209.1(GAS8):c.994C>T (p.Gln332Ter)	GAS8	16	90040357	Ciliary dyskinesia, primary, 33
NM_173494.1(PIH1D3):c.355C>T (p.Gln119Ter)	PIH1D3	X	107222767	CILIARY DYSKINESIA, PRIMARY, 36, X-LINKED
NM_001277115.1(DNAH11):c.8533C>T (p.Arg2845Ter)	DNAH11	7	21748602	Ciliary dyskinesia, primary, 7
NM_001277115.1(DNAH11):c.7772C>T (p.Pro2591Leu)	DNAH11	7	21738827	Ciliary dyskinesia, primary, 7
NM_001277115.1(DNAH11):c.8746C>T (p.Gln2916Ter)	DNAH11	7	21749750	Ciliary dyskinesia, primary, 7
NM_001277115.1(DNAH11):c.9706C>T (p.Arg3236Ter)	DNAH11	7	21786732	Ciliary dyskinesia, primary, 7
NM_001277115.1(DNAH11):c.8698C>T (p.Arg2900Ter)	DNAH11	7	21749702	Ciliary dyskinesia, primary, 7;Primary ciliary dyskinesia
NM_001277115.1(DNAH11):c.6727C>T (p.Arg2243Ter)	DNAH11	7	21710596	Ciliary dyskinesia, primary, 7;Primary ciliary dyskinesia
NM_023036.4(DNAI2):c.787C>T (p.Arg263Ter)	DNAI2	17	74299780	Ciliary dyskinesia, primary, 9
NM_023036.4(DNAI2):c.1304G>A (p.Trp435Ter)	DNAI2	17	74309345	Ciliary dyskinesia, primary, 9;Primary ciliary dyskinesia
NM_014251.2(SLC25A13):c.1177+1G>A	SLC25A13	7	96184276	Citrin deficiency;Citrullinemia type II;Neonatal intrahepatic cholestasis caused by citrin deficiency
NM_000050.4(ASS1):c.40G>A (p.Gly14Ser)	ASS1	9	130452268	Citrullinemia type I
NM_000050.4(ASS1):c.970G>A (p.Gly324Ser)	ASS1	9	130489464	Citrullinemia type I
NM_000050.4(ASS1):c.910C>T (p.Arg304Trp)	ASS1	9	130489404	Citrullinemia type I
NM_000050.4(ASS1):c.53C>T (p.Ser18Leu)	ASS1	9	130452281	Citrullinemia type I
NM_000050.4(ASS1):c.256C>T (p.Arg86Cys)	ASS1	9	130458482	Citrullinemia type I
NM_000050.4(ASS1):c.835C>T (p.Arg279Ter)	ASS1	9	130480446	Citrullinemia type I
NM_000050.4(ASS1):c.793C>T (p.Arg265Cys)	ASS1	9	130480404	Citrullinemia type I
NM_000050.4(ASS1):c.794G>A (p.Arg265His)	ASS1	9	130480405	Citrullinemia type I
NM_000050.4(ASS1):c.836G>A (p.Arg279Gln)	ASS1	9	130480447	Citrullinemia type I
NM_000050.4(ASS1):c.571G>A (p.Glu191Lys)	ASS1	9	130471489	Citrullinemia type I
NM_000050.4(ASS1):c.257G>A (p.Arg86His)	ASS1	9	130458483	Citrullinemia type I
NM_000050.4(ASS1):c.773+49C>T	ASS1	9	130479849	Citrullinemia type I

NM_000050.4(ASS1):c.851C>T (p.Thr284Ile)	ASS1	9	130489345	Citrullinemia type I
NM_000050.4(ASS1):c.970+5G>A	ASS1	9	130489469	Citrullinemia type I
NM_000050.4(ASS1):c.539G>A (p.Ser180Asn)	ASS1	9	130470877	Citrullinemia type I;not provided
NM_000050.4(ASS1):c.1087C>T (p.Arg363Trp)	ASS1	9	130494983	Citrullinemia type I;not provided
NM_000050.4(ASS1):c.1168G>A (p.Gly390Arg)	ASS1	9	130499545	Citrullinemia type I;not provided
NM_000050.4(ASS1):c.787G>A (p.Val263Met)	ASS1	9	130480398	Citrullinemia type I;not provided
NM_014251.2(SLC25A13):c.1311+1G>A	SLC25A13	7	96170044	Citrullinemia type II
NM_014251.2(SLC25A13):c.1763G>A (p.Arg588Gln)	SLC25A13	7	96121733	Citrullinemia type II
NM_001830.3(CLCN4):c.1601C>T (p.Ser534Leu)	CLCN4	X	10213705	CLCN4-related disorder
NM_001830.3(CLCN4):c.2152C>T (p.Arg718Trp)	CLCN4	X	10220837	CLCN4-related disorder
NM_001830.3(CLCN4):c.823G>A (p.Val275Met)	CLCN4	X	10206756	CLCN4-related disorder
NM_001830.3(CLCN4):c.1389+5G>A	CLCN4	X	10208595	CLCN4-related disorder
NM_001830.3(CLCN4):c.1664C>T (p.Ala555Val)	CLCN4	X	10213768	CLCN4-related disorder;not provided
NM_016828.2(OGG1):c.137G>A (p.Arg46Gln)	OGG1	3	9750423	Clear cell carcinoma of kidney
NM_203285.1(NECTIN1):c.554G>A (p.Trp185Ter)	NECTIN1	11	119677734	Cleft lip/palate-ectodermal dysplasia syndrome;Orofacial cleft 7
NM_001109878.1(TBX22):c.779C>T (p.Thr260Met)	TBX22	X	80026849	Cleft palate with ankyloglossia
NM_001009999.2(KDM1A):c.1207G>A (p.Glu403Lys)	KDM1A	1	23068566	Cleft palate, psychomotor retardation, and distinctive facial features
NM_001110556.1(FLNA):c.620C>T (p.Pro207Leu)	FLNA	X	154367844	Cleft palate;Conductive hearing impairment;Oto-palato-digital syndrome, type I;Short stature
NM_006147.3(IRF6):c.16C>T (p.Arg6Cys)	IRF6	1	209801398	Cleft palate;Van der Woude syndrome;not provided
NM_001024630.3(RUNX2):c.891G>A (p.Trp297Ter)	RUNX2	6	45512277	Cleidocranial dysostosis
NM_001024630.3(RUNX2):c.572G>A (p.Ser191Asn)	RUNX2	6	45432011	Cleidocranial dysostosis
NM_001024630.3(RUNX2):c.674G>A (p.Arg225Gln)	RUNX2	6	45438040	Cleidocranial dysostosis
NM_001024630.3(RUNX2):c.673C>T (p.Arg225Trp)	RUNX2	6	45438039	Cleidocranial dysostosis
NM_001024630.3(RUNX2):c.1171C>T (p.Arg391Ter)	RUNX2	6	45546910	Cleidocranial dysostosis
NM_001015051.3(RUNX2):c.476G>A (p.Gly159Asp)	RUNX2	6	45431915	Cleidocranial dysostosis
NM_015338.5(ASXL1):c.2773C>T (p.Gln925Ter)	ASXL1	20	32435485	C-like syndrome
NM_015338.5(ASXL1):c.2197C>T (p.Gln733Ter)	ASXL1	20	32434909	C-like syndrome
NM_015338.5(ASXL1):c.2893C>T (p.Arg965Ter)	ASXL1	20	32435605	C-like syndrome;not provided
NM_153704.5(TMEM67):c.312+5G>A	TMEM67	8	93755871	COACH syndrome
NM_015272.4(RPGRIP1L):c.2413C>T (p.Arg805Ter)	RPGRIP1L	16	53645895	COACH syndrome;Joubert syndrome 7
NM_001080522.2(CC2D2A):c.3347C>T (p.Thr1116Met)	CC2D2A	4	15567735	COACH syndrome;Joubert syndrome 9
NM_001080522.2(CC2D2A):c.3145C>T (p.Arg1049Ter)	CC2D2A	4	15563485	COACH syndrome;Joubert syndrome;Joubert syndrome 9/15, digenic
NM_000124.3(ERCC6):c.1550G>A (p.Trp517Ter)	ERCC6	10	49500673	Cockayne syndrome B
NM_000124.3(ERCC6):c.1357C>T (p.Arg453Ter)	ERCC6	10	49524073	Cockayne syndrome B
NM_000124.3(ERCC6):c.3904C>T (p.Gln1302Ter)	ERCC6	10	49461431	Cockayne syndrome B
NM_000124.3(ERCC6):c.2008C>T (p.Arg670Trp)	ERCC6	10	49482848	Cockayne syndrome B
NM_000124.3(ERCC6):c.2203C>T (p.Arg735Ter)	ERCC6	10	49478437	Cockayne syndrome B;DE SANCTIS-CACCHIONE SYNDROME;ERCC6-Related Disorders
NM_000124.3(ERCC6):c.2167C>T (p.Gln723Ter)	ERCC6	10	49482689	Cockayne syndrome B;not provided
NM_000124.3(ERCC6):c.466C>T (p.Gln156Ter)	ERCC6	10	49530797	Cockayne syndrome B;not provided
NM_000082.3(ERCC8):c.479C>T (p.Ala160Val)	ERCC8	5	60904794	Cockayne syndrome type A;not provided
NM_001278.4(CHUK):c.1264C>T (p.Gln422Ter)	CHUK	10	100205167	Cocoon syndrome
NM_004793.3(LONP1):c.2026C>T (p.Pro676Ser)	LONP1	19	5694889	CODAS syndrome
NM_004793.3(LONP1):c.2171C>T (p.Ala724Val)	LONP1	19	5694536	CODAS syndrome
NM_015697.7(COQ2):c.905C>T (p.Ala302Val)	COQ2	4	83269867	Coenzyme Q10 deficiency, primary
NM_020381.3(PDSS2):c.964C>T (p.Gln322Ter)	PDSS2	6	107210483	Coenzyme Q10 deficiency, primary, 3

NM_020381.3(PDSS2):c.1145C>T (p.Ser382Leu)	PDSS2	6	107154674	Coenzyme Q10 deficiency, primary, 3	
NM_020247.4(COQ8A):c.1651G>A (p.Glu551Lys)	COQ8A	1	226985332	Coenzyme Q10 deficiency, primary, 4	
NM_020247.4(COQ8A):c.637C>T (p.Arg213Trp)	COQ8A	1	226965719	Coenzyme Q10 deficiency, primary, 4	
NM_020247.4(COQ8A):c.815G>A (p.Gly272Asp)	COQ8A	1	226982111	Coenzyme Q10 deficiency, primary, 4	
NM_020247.4(COQ8A):c.1645G>A (p.Gly549Ser)	COQ8A	1	226985326	Coenzyme Q10 deficiency, primary, 4	
NM_020247.4(COQ8A):c.1844G>A (p.Gly615Asp)	COQ8A	1	226986637	Coenzyme Q10 deficiency, primary, 4	
NM_020247.4(COQ8A):c.1042C>T (p.Arg348Ter)	COQ8A	1	226982996	Coenzyme Q10 deficiency, primary, 4;not provided	
NM_020247.4(COQ8A):c.895C>T (p.Arg299Trp)	COQ8A	1	226982719	Coenzyme Q10 deficiency, primary, 4;not provided	
NM_020312.3(COQ9):c.730C>T (p.Arg244Ter)	COQ9	16	57459583	Coenzyme Q10 deficiency, primary, 5	
NM_182476.2(COQ6):c.484C>T (p.Arg162Ter)	COQ6	14	73958149	Coenzyme Q10 deficiency, primary, 6	
NM_182476.2(COQ6):c.564G>A (p.Trp188Ter)	COQ6	14	73958229	Coenzyme Q10 deficiency, primary, 6	
NM_182476.2(COQ6):c.763G>A (p.Gly255Arg)	COQ6	14	73959204	Coenzyme Q10 deficiency, primary, 6	
NM_182476.2(COQ6):c.1341G>A (p.Trp447Ter)	COQ6	14	73961867	Coenzyme Q10 deficiency, primary, 6	
NM_016035.4(COQ4):c.421C>T (p.Arg141Ter)	COQ4	9	128332171	Coenzyme Q10 deficiency, primary, 7	
NM_016035.4(COQ4):c.190C>T (p.Pro64Ser)	COQ4	9	128323135	Coenzyme Q10 deficiency, primary, 7	
NM_016035.4(COQ4):c.718C>T (p.Arg240Cys)	COQ4	9	128333565	Coenzyme Q10 deficiency, primary, 7;not provided	
NM_015697.7(COQ2):c.590G>A (p.Arg197His)	COQ2	4	83273598	Coenzyme Q10 deficiency, primary;Coenzyme Q10 deficiency, primary 1	
NM_015697.7(COQ2):c.437G>A (p.Ser146Asn)	COQ2	4	83279081	Coenzyme Q10 deficiency, primary;Coenzyme Q10 deficiency, primary 1	
NM_015697.7(COQ2):c.1159C>T (p.Arg387Ter)	COQ2	4	83264306	Coenzyme Q10 deficiency, primary;Shy-Drager syndrome	
NM_020732.3(ARID1B):c.4102C>T (p.Gln1368Ter)	ARID1B	6	157198899	Coffin Siris/Intellectual Disability	
NM_004586.2(RPS6KA3):c.340C>T (p.Arg114Trp)	RPS6KA3	X	20195131	Coffin-Lowry syndrome	
NM_004586.2(RPS6KA3):c.2065C>T (p.Gln689Ter)	RPS6KA3	X	20156144	Coffin-Lowry syndrome	
NM_004586.2(RPS6KA3):c.913C>T (p.Arg305Ter)	RPS6KA3	X	20177017	Coffin-Lowry syndrome	
NM_004586.2(RPS6KA3):c.2186G>A (p.Arg729Gln)	RPS6KA3	X	20155435	Coffin-Lowry syndrome;not provided	
NM_020732.3(ARID1B):c.3919C>T (p.Gln1307Ter)	ARID1B	6	157196221	Coffin-Siris syndrome 1	
NM_020732.3(ARID1B):c.1903C>T (p.Gln635Ter)	ARID1B	6	156901502	Coffin-Siris syndrome 1	
NM_020732.3(ARID1B):c.3304C>T (p.Arg1102Ter)	ARID1B	6	157181137	Coffin-Siris syndrome 1;not provided	
NM_020732.3(ARID1B):c.3223C>T (p.Arg1075Ter)	ARID1B	6	157181056	Coffin-Siris syndrome 1;not provided	
NM_017890.4(VPS13B):c.7051C>T (p.Arg2351Ter)	VPS13B	8	99720973	Cohen syndrome	
NM_017890.4(VPS13B):c.2911C>T (p.Arg971Ter)	VPS13B	8	99384294	Cohen syndrome	
NM_017890.4(VPS13B):c.7934G>A (p.Gly2645Asp)	VPS13B	8	99784394	Cohen syndrome	
NM_017890.4(VPS13B):c.10888C>T (p.Gln3630Ter)	VPS13B	8	99854202	Cohen syndrome	
NM_017890.4(VPS13B):c.5827C>T (p.Arg1943Ter)	VPS13B	8	99642342	Cohen syndrome	
NM_017890.4(VPS13B):c.8990G>A (p.Trp2997Ter)	VPS13B	8	99820043	Cohen syndrome	
NM_017890.4(VPS13B):c.2158C>T (p.Gln720Ter)	VPS13B	8	99156693	Cohen syndrome	
NM_017890.4(VPS13B):c.11314C>T (p.Gln3772Ter)	VPS13B	8	99868312	Cohen syndrome;Intellectual disability;Microcephaly;Neutropenia;Progressive visual loss;Recurrent aphthous stomatitis;Retinal dystrophy;Short foot;Short stature;Small hand	
NM_017890.4(VPS13B):c.10946G>A (p.Trp3649Ter)	VPS13B	8	99859307	Cohen syndrome;not provided	
NM_017890.4(VPS13B):c.1219C>T (p.Gln407Ter)	VPS13B	8	99134644	Cohen syndrome;not provided	
NM_017890.4(VPS13B):c.2074C>T (p.Arg692Ter)	VPS13B	8	99156609	Cohen syndrome;not provided	
NM_017890.4(VPS13B):c.2889G>A (p.Trp963Ter)	VPS13B	8	99384272	Cohen syndrome;not provided	
NM_017890.4(VPS13B):c.3427C>T (p.Arg1143Ter)	VPS13B	8	99442617	Cohen syndrome;not provided	
NM_017890.4(VPS13B):c.6732+1G>A	VPS13B	8	99717374	Cohen syndrome;not provided	
NM_017890.4(VPS13B):c.4923G>A (p.Trp1641Ter)	VPS13B	8	99556552	Cohen syndrome;not provided	
NM_017890.4(VPS13B):c.3598C>T (p.Arg1200Ter)	VPS13B	8	99467566	Cohen syndrome;not provided	

NM_004750.4(CRLF1):c.829C>T (p.Arg277Ter)	CRLF1	19	18596918	Cold-induced sweating syndrome 1
NM_004750.4(CRLF1):c.397+1G>A	CRLF1	19	18599564	Cold-induced sweating syndrome 1
NM_004750.4(CRLF1):c.413C>T (p.Pro138Leu)	CRLF1	19	18598886	Cold-induced sweating syndrome 1
NM_004750.4(CRLF1):c.538C>T (p.Gln180Ter)	CRLF1	19	18598591	Cold-induced sweating syndrome 1
NM_004750.4(CRLF1):c.935G>A (p.Arg312His)	CRLF1	19	18596711	Cold-induced sweating syndrome 1
NM_001031710.2(KLHL7):c.1115G>A (p.Arg372Gln)	KLHL7	7	23165876	Cold-induced sweating syndrome 3
NM_001031710.2(KLHL7):c.1258C>T (p.Arg420Cys)	KLHL7	7	23167916	Cold-induced sweating syndrome 3
NM_006208.2(ENPP1):c.530G>A (p.Cys177Tyr)	ENPP1	6	131851241	Cole disease
NM_014822.3(SEC24D):c.3044C>T (p.Ser1015Phe)	SEC24D	4	118723570	Cole-Carpenter syndrome 2
NM_014822.3(SEC24D):c.613C>T (p.Gln205Ter)	SEC24D	4	118815511	Cole-Carpenter syndrome 2
NM_000280.4(PAX6):c.202C>T (p.Pro68Ser)	PAX6	11	31801716	Coloboma of optic disc
NM_000248.3(MITF):c.635-1G>A	MITF	3	69956454	COLOBOMA, OSTEOPETROSIS, MICROPHTHALMIA, MACROCEPHALY, ALBINISM, AND DEAFNESS;Waardenburg syndrome type 2A
NM_198291.2(SRC):c.1591C>T (p.Gln531Ter)	SRC	20	37403359	Colon cancer, advanced;Neoplasm
NM_017763.5(RNF43):c.394C>T (p.Arg132Ter)	RNF43	17	58363582	Colon Serrated Polyposis;SESSILE SERRATED POLYPOSIS CANCER SYNDROME
NM_002539.2(ODC1):c.-128+109G>A	ODC1	2	10448012	Colonic adenoma recurrence, reduced risk of
NM_000513.2(OPN1MW):c.989G>A (p.Arg330Gln)	OPN1MW	X	154195934	Colorblindness, partial, deutan series
NM_024642.4(GALNT12):c.1472C>T (p.Thr491Met)	GALNT12	9	98845990	Colorectal cancer 1
NM_004336.4(BUB1):c.422+1G>A	BUB1	2	110672660	Colorectal cancer with chromosomal instability
NM_000535.6(PMS2):c.943C>T (p.Arg315Ter)	PMS2	7	5992018	Colorectal cancer, non-polyposis;Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000038.5(APC):c.562C>T (p.Gln188Ter)	APC	5	112780820	Colorectal cancer, susceptibility to
NM_006231.3(POLE):c.1420G>A (p.Val474Ile)	POLE	12	132673217	Colorectal cancer, susceptibility to, 12
NM_000038.5(APC):c.904C>T (p.Arg302Ter)	APC	5	112815564	Colorectal cancer, susceptibility to;Familial adenomatous polyposis 1;Gardner syndrome;Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.1660C>T (p.Arg554Ter)	APC	5	112828889	Colorectal cancer, susceptibility to;Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome;not provided
NM_053056.2(CCND1):c.723G>A (p.Pro241=)	CCND1	11	69648142	Colorectal cancer, susceptibility to;Multiple myeloma, translocation 11,14 type
NM_005359.5(SMAD4):c.1051G>A (p.Asp351Asn)	SMAD4	18	51065518	Colorectal Neoplasms
NM_005343.3(HRAS):c.38G>A (p.Gly13Asp)	HRAS	11	534285	Colorectal Neoplasms;Costello syndrome;not provided
NM_000038.5(APC):c.4348C>T (p.Arg1450Ter)	APC	5	112839942	Colorectal Neoplasms;Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.2626C>T (p.Arg876Ter)	APC	5	112838220	Colorectal Neoplasms;Familial adenomatous polyposis 1;not provided
NM_004958.3(MTOR):c.6644C>T (p.Ser2215Phe)	MTOR	1	11124516	Colorectal Neoplasms;Glioblastoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Papillary renal cell carcinoma, sporadic;Renal cell carcinoma;Uterine cervical neoplasms;not provided
NM_000546.5(TP53):c.916C>T (p.Arg306Ter)	TP53	17	7673704	Colorectal Neoplasms;Head and Neck Neoplasms;Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;not provided
NM_033360.3(KRAS):c.34G>A (p.Gly12Ser)	KRAS	12	25245351	Colorectal Neoplasms;Juvenile myelomonocytic leukemia;Neoplasm of ovary;Neoplasm of stomach;Neoplasm of the thyroid gland;Non-small cell lung cancer
NM_002755.3(MAP2K1):c.370C>T (p.Pro124Ser)	MAP2K1	15	66436824	Colorectal Neoplasms;Malignant melanoma;Malignant melanoma of skin;Malignant neoplasm of body of uterus;Transitional cell carcinoma of the bladder;not provided
NM_006218.3(PIK3CA):c.1645G>A (p.Asp549Asn)	PIK3CA	3	179218315	Colorectal Neoplasms;Neoplasm of breast
NM_004985.4(KRAS):c.437C>T (p.Ala146Val)	KRAS	12	25225627	Colorectal Neoplasms;Neoplasm of the thyroid gland;Non-small cell lung cancer
NM_033360.3(KRAS):c.37G>A (p.Gly13Ser)	KRAS	12	25245348	Colorectal Neoplasms;Neoplasm of the thyroid gland;Non-small cell lung cancer

NM_005228.4(EGFR):c.2155G>A (p.Gly719Ser)	EGFR	7	55174014	Colorectal Neoplasms;Non-small cell lung cancer;Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, somatic;Tyrosine kinase inhibitor response
NM_000448.2(RAG1):c.940C>T (p.Arg314Trp)	RAG1	11	36574244	Combined cellular and humoral immune defects with granulomas
NM_000448.2(RAG1):c.2923C>T (p.Arg975Trp)	RAG1	11	36576227	Combined cellular and humoral immune defects with granulomas
NM_000448.2(RAG1):c.2210G>A (p.Arg737His)	RAG1	11	36575514	Combined cellular and humoral immune defects with granulomas;Histiocytic medullary reticulosis
NM_000448.2(RAG1):c.2333G>A (p.Arg778Gln)	RAG1	11	36575637	Combined cellular and humoral immune defects with granulomas;not provided
NM_005984.4(SLC25A1):c.844C>T (p.Arg282Cys)	SLC25A1	22	19176222	Combined d-2- and l-2-hydroxyglutaric aciduria
NM_005984.4(SLC25A1):c.845G>A (p.Arg282His)	SLC25A1	22	19176221	Combined d-2- and l-2-hydroxyglutaric aciduria
NM_005984.4(SLC25A1):c.389G>A (p.Gly130Asp)	SLC25A1	22	19177779	Combined d-2- and l-2-hydroxyglutaric aciduria
NM_005984.4(SLC25A1):c.821C>T (p.Ala274Val)	SLC25A1	22	19176421	Combined d-2- and l-2-hydroxyglutaric aciduria
NM_000308.3(CTSA):c.323C>T (p.Ser108Leu)	CTSA	20	45891990	Combined deficiency of sialidase AND beta galactosidase
NM_000308.3(CTSA):c.448G>A (p.Val150Met)	CTSA	20	45892434	Combined deficiency of sialidase AND beta galactosidase;Galactosialidosis, early infantile
NM_000206.2(IL2RG):c.664C>T (p.Arg222Cys)	IL2RG	X	71109321	Combined immunodeficiency, X-linked
NM_174917.4(ACSF3):c.1567C>T (p.Arg523Ter)	ACSF3	16	89146003	Combined malonic and methylmalonic aciduria
NM_174917.4(ACSF3):c.1411C>T (p.Arg471Trp)	ACSF3	16	89145311	Combined malonic and methylmalonic aciduria
NM_174917.4(ACSF3):c.1412G>A (p.Arg471Gln)	ACSF3	16	89145312	Combined malonic and methylmalonic aciduria
NM_174917.4(ACSF3):c.1073C>T (p.Thr358Ile)	ACSF3	16	89114434	Combined malonic and methylmalonic aciduria
NM_174917.4(ACSF3):c.728C>T (p.Pro243Leu)	ACSF3	16	89102665	Combined malonic and methylmalonic aciduria
NM_174917.4(ACSF3):c.1672C>T (p.Arg558Trp)	ACSF3	16	89154148	Combined malonic and methylmalonic aciduria;not provided
NM_024996.5(GFM1):c.139C>T (p.Arg47Ter)	GFM1	3	158645686	Combined oxidative phosphorylation deficiency 1
NM_024996.5(GFM1):c.748C>T (p.Arg250Trp)	GFM1	3	158652154	Combined oxidative phosphorylation deficiency 1
NM_133645.2(MTO1):c.1357G>A (p.Ala453Thr)	MTO1	6	73482061	Combined oxidative phosphorylation deficiency 10
NM_133645.2(MTO1):c.1307C>T (p.Thr436Ile)	MTO1	6	73480777	Combined oxidative phosphorylation deficiency 10
NM_133645.2(MTO1):c.1505G>A (p.Arg502His)	MTO1	6	73482209	Combined oxidative phosphorylation deficiency 10;not provided
NM_017909.3(RMND1):c.1250G>A (p.Arg417Gln)	RMND1	6	151405787	Combined oxidative phosphorylation deficiency 11;Mitochondrial diseases
NM_001083614.1(EARS2):c.610G>A (p.Gly204Ser)	EARS2	16	23535236	Combined oxidative phosphorylation deficiency 12
NM_001083614.1(EARS2):c.286G>A (p.Glu96Lys)	EARS2	16	23552158	Combined oxidative phosphorylation deficiency 12
NM_001083614.1(EARS2):c.500G>A (p.Cys167Tyr)	EARS2	16	23535346	Combined oxidative phosphorylation deficiency 12
NM_001083614.1(EARS2):c.322C>T (p.Arg108Trp)	EARS2	16	23544677	Combined oxidative phosphorylation deficiency 12;not provided
NM_001083614.1(EARS2):c.328G>A (p.Gly110Ser)	EARS2	16	23544671	Combined oxidative phosphorylation deficiency 12;not provided
NM_139242.3(MTFMT):c.382C>T (p.Arg128Ter)	MTFMT	15	65026868	Combined oxidative phosphorylation deficiency 15
NM_139242.3(MTFMT):c.374C>T (p.Ser125Leu)	MTFMT	15	65026876	Combined oxidative phosphorylation deficiency 15
NM_139242.3(MTFMT):c.452C>T (p.Pro151Leu)	MTFMT	15	65023762	Combined oxidative phosphorylation deficiency 15
NM_139242.3(MTFMT):c.878G>A (p.Ser293Asn)	MTFMT	15	65006127	Combined oxidative phosphorylation deficiency 15
NM_139242.3(MTFMT):c.73C>T (p.Gln25Ter)	MTFMT	15	65029541	Combined oxidative phosphorylation deficiency 15
NM_139242.3(MTFMT):c.994C>T (p.Arg332Ter)	MTFMT	15	65003238	Combined oxidative phosphorylation deficiency 15;Leigh syndrome due to mitochondrial complex I deficiency;not provided
NM_018127.6(ELAC2):c.1559C>T (p.Thr520Ile)	ELAC2	17	12996647	Combined oxidative phosphorylation deficiency 17
NM_018127.6(ELAC2):c.1267C>T (p.Leu423Phe)	ELAC2	17	13002311	Combined oxidative phosphorylation deficiency 17
NM_213649.1(SFXN4):c.471+1G>A	SFXN4	10	119157870	Combined oxidative phosphorylation deficiency 18
NM_016065.3(MRPS16):c.331C>T (p.Arg111Ter)	MRPS16	10	73250935	Combined oxidative phosphorylation deficiency 2
NM_001167734.1(VARS2):c.1135G>A (p.Ala379Thr)	VARS2	6	30918886	Combined oxidative phosphorylation deficiency 20
NM_001167734.1(VARS2):c.1100C>T (p.Thr367Ile)	VARS2	6	30918851	Combined oxidative phosphorylation deficiency 20
NM_025150.4(TARS2):c.845C>T (p.Pro282Leu)	TARS2	1	150496552	Combined oxidative phosphorylation deficiency 21
NM_001195422.1(GTPBP3):c.1441G>A (p.Glu481Lys)	GTPBP3	19	17341599	Combined oxidative phosphorylation deficiency 23

NM_024678.5(NARS2):c.641C>T (p.Pro214Leu)	NARS2	11	78528890	Combined oxidative phosphorylation deficiency 24
NM_138395.3(MARS2):c.550C>T (p.Gln184Ter)	MARS2	2	197705955	Combined oxidative phosphorylation deficiency 25
NM_138395.3(MARS2):c.424C>T (p.Arg142Trp)	MARS2	2	197705829	Combined oxidative phosphorylation deficiency 25
NM_020810.3(TRMT5):c.872G>A (p.Arg291His)	TRMT5	14	60976047	Combined oxidative phosphorylation deficiency 26
NM_024537.3(CARS2):c.655G>A (p.Ala219Thr)	CARS2	13	110683051	Combined oxidative phosphorylation deficiency 27
NM_173471.3(SLC25A26):c.305C>T (p.Ala102Val)	SLC25A26	3	66262055	Combined oxidative phosphorylation deficiency 28
NM_173471.3(SLC25A26):c.596C>T (p.Pro199Leu)	SLC25A26	3	66369505	Combined oxidative phosphorylation deficiency 28
NM_173471.3(SLC25A26):c.33+1G>A	SLC25A26	3	66221128	Combined oxidative phosphorylation deficiency 28
NM_012473.3(TXN2):c.71G>A (p.Trp24Ter)	TXN2	22	36480767	Combined oxidative phosphorylation deficiency 29
NM_001172696.1(TSFM):c.997C>T (p.Arg333Trp)	TSFM	12	57796539	Combined oxidative phosphorylation deficiency 3
NM_005726.5(TSFM):c.944G>A (p.Cys315Tyr)	TSFM	12	57796549	Combined oxidative phosphorylation deficiency 3
NM_005726.5(TSFM):c.856C>T (p.Gln286Ter)	TSFM	12	57796461	Combined oxidative phosphorylation deficiency 3;Primary dilated cardiomyopathy
NM_005932.3(MIPEP):c.916C>T (p.Leu306Phe)	MIPEP	13	23869319	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 31
NM_003321.4(TUFM):c.1016G>A (p.Arg339Gln)	TUFM	16	28844008	Combined oxidative phosphorylation deficiency 4
NM_020191.2(MRPS22):c.509G>A (p.Arg170His)	MRPS22	3	139350183	Combined oxidative phosphorylation deficiency 5
NM_020745.3(AARS2):c.1774C>T (p.Arg592Trp)	AARS2	6	44304512	Combined oxidative phosphorylation deficiency 8;not provided
NM_000102.3(CYP17A1):c.1039C>T (p.Arg347Cys)	CYP17A1	10	102832611	Combined partial 17-alpha-hydroxylase/17,20-lyase deficiency
NM_001077494.3(NFKB2):c.2557C>T (p.Arg853Ter)	NFKB2	10	102402138	Common variable immunodeficiency 1;Common variable immunodeficiency 10
NM_001077494.3(NFKB2):c.2600C>T (p.Ala867Val)	NFKB2	10	102402273	Common variable immunodeficiency 10
NM_001006658.2(CR2):c.2297G>A (p.Trp766Ter)	CR2	1	207474297	Common variable immunodeficiency 7
NM_006726.4(LRBA):c.5047C>T (p.Arg1683Ter)	LRBA	4	150828304	Common variable immunodeficiency 8, with autoimmunity
NM_006726.4(LRBA):c.2032C>T (p.Gln678Ter)	LRBA	4	150896429	Common variable immunodeficiency 8, with autoimmunity
NM_001734.3(C1S):c.1600C>T (p.Arg534Trp)	C1S	12	7070184	Complement 1s deficiency
NM_000062.2(SERPING1):c.1394C>T (p.Ala465Val)	SERPING1	11	57614472	Complement component 4, partial deficiency of, due to dysfunctional c1 inhibitor
NM_000066.3(C8B):c.1282C>T (p.Arg428Ter)	C8B	1	56940965	Complement component 8 deficiency type 2
NM_000066.3(C8B):c.820C>T (p.Arg274Ter)	C8B	1	56949599	Complement component 8 deficiency type 2
NM_000066.3(C8B):c.361C>T (p.Arg121Ter)	C8B	1	56956799	Complement component 8 deficiency type 2
NM_000066.3(C8B):c.271C>T (p.Gln91Ter)	C8B	1	56956889	Complement component 8 deficiency type 2
NM_001737.4(C9):c.346C>T (p.Arg116Ter)	C9	5	39341276	Complement component 9 deficiency
NM_001710.5(CFB):c.766C>T (p.Gln256Ter)	CFB	6	31947950	Complement factor B deficiency
NM_000102.3(CYP17A1):c.51G>A (p.Trp17Ter)	CYP17A1	10	102837311	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency
NM_000102.3(CYP17A1):c.1084C>T (p.Arg362Cys)	CYP17A1	10	102832566	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency
NM_000102.3(CYP17A1):c.1283C>T (p.Pro428Leu)	CYP17A1	10	102830946	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency
NM_000102.3(CYP17A1):c.287G>A (p.Arg96Gln)	CYP17A1	10	102837075	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency
NM_000102.3(CYP17A1):c.374G>A (p.Arg125Gln)	CYP17A1	10	102835316	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency
NM_000102.3(CYP17A1):c.1247G>A (p.Arg416His)	CYP17A1	10	102830982	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency
NM_000102.3(CYP17A1):c.286C>T (p.Arg96Trp)	CYP17A1	10	102837076	Complete combined 17-alpha-hydroxylase/17,20-lyase deficiency;not provided
NM_000409.4(GUCA1A):c.451C>T (p.Leu151Phe)	GUCA1A	6	42179248	Cone dystrophy 3
NM_000409.4(GUCA1A):c.250C>T (p.Leu84Phe)	GUCA1A	6	42178328	Cone dystrophy 3
NM_006204.3(PDE6C):c.85C>T (p.Arg29Trp)	PDE6C	10	93612810	Cone dystrophy 4
NM_020061.5(OPN1LW):c.739C>T (p.Arg247Ter)	OPN1LW	X	154154734	Cone monochromatism
NM_020366.3(RPGRIP1):c.154C>T (p.Arg52Ter)	RPGRIP1	14	21294745	Cone-rod dystrophy 13
NM_004249.3(RAB28):c.565C>T (p.Gln189Ter)	RAB28	4	13376553	Cone-rod dystrophy 18
NM_001017979.2(RAB28):c.409C>T (p.Arg137Ter)	RAB28	4	13381577	Cone-rod dystrophy 18
NM_015072.4(TTLL5):c.3354G>A (p.Trp1118Ter)	TTLL5	14	75863694	Cone-rod dystrophy 19

NM_015072.4(TTLL5):c.1627G>A (p.Glu543Lys)	TTLL5	14	75764691	Cone-rod dystrophy 19
NM_000554.5(CRX):c.121C>T (p.Arg41Trp)	CRX	19	47836263	Cone-rod dystrophy 2;not provided
NM_000554.5(CRX):c.122G>A (p.Arg41Gln)	CRX	19	47836264	Cone-rod dystrophy 2;not provided
NM_178454.5(DRAM2):c.494G>A (p.Trp165Ter)	DRAM2	1	111120539	Cone-rod dystrophy 21;Retinal dystrophy
NM_178454.5(DRAM2):c.131G>A (p.Ser44Asn)	DRAM2	1	111131424	Cone-rod dystrophy 21;Retinal dystrophy
NM_000350.2(ABCA4):c.763C>T (p.Arg255Cys)	ABCA4	1	94098799	Cone-rod dystrophy 3
NM_000350.2(ABCA4):c.161G>A (p.Cys54Tyr)	ABCA4	1	94111579	Cone-rod dystrophy 3;Retinitis pigmentosa 19;Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.5714+5G>A	ABCA4	1	94010795	Cone-rod dystrophy 3;Retinitis pigmentosa 19;Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.880C>T (p.Gln294Ter)	ABCA4	1	94080697	Cone-rod dystrophy 3;Stargardt disease 1
NM_000350.2(ABCA4):c.6079C>T (p.Leu2027Phe)	ABCA4	1	94005509	Cone-rod dystrophy 3;Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.3113C>T (p.Ala1038Val)	ABCA4	1	94043413	Cone-rod dystrophy 3;Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.634C>T (p.Arg212Cys)	ABCA4	1	94098928	Cone-rod dystrophy 3;Stargardt disease 1;not provided
NM_000180.3(GUCY2D):c.2512C>T (p.Arg838Cys)	GUCY2D	17	8014700	Cone-rod dystrophy 6;not provided
NM_000180.3(GUCY2D):c.2513G>A (p.Arg838His)	GUCY2D	17	8014701	Cone-rod dystrophy 6;not provided
NM_014989.5(RIMS1):c.2459G>A (p.Arg820His)	RIMS1	6	72251007	Cone-rod dystrophy 7
NM_003816.2(ADAM9):c.1130+1G>A	ADAM9	8	39026811	Cone-rod dystrophy 9
NM_003816.2(ADAM9):c.766C>T (p.Arg256Ter)	ADAM9	8	39023177	Cone-rod dystrophy 9
NM_003816.2(ADAM9):c.490C>T (p.Arg164Ter)	ADAM9	8	39017298	Cone-rod dystrophy 9;not provided
NM_020184.3(CNNM4):c.707G>A (p.Arg236Gln)	CNNM4	2	96761706	Cone-rod dystrophy amelogenesis imperfecta
NM_020184.3(CNNM4):c.1690C>T (p.Gln564Ter)	CNNM4	2	96799065	Cone-rod dystrophy amelogenesis imperfecta
NM_020184.3(CNNM4):c.2149C>T (p.Gln717Ter)	CNNM4	2	96809338	Cone-rod dystrophy amelogenesis imperfecta
NM_001098802.2(CEP78):c.499+5G>A	CEP78	9	78240369	CONE-ROD DYSTROPHY AND HEARING LOSS
NM_032171.2(CEP78):c.893-1G>A	CEP78	9	78248290	CONE-ROD DYSTROPHY AND HEARING LOSS
NM_032171.2(CEP78):c.1254+5G>A	CEP78	9	78253282	CONE-ROD DYSTROPHY AND HEARING LOSS
NM_005183.3(CACNA1F):c.2542G>A (p.Gly848Ser)	CACNA1F	X	49219668	Cone-rod dystrophy X-linked 3
NM_005183.3(CACNA1F):c.244C>T (p.Arg82Ter)	CACNA1F	X	49231709	Cone-rod dystrophy X-linked 3;Congenital stationary night blindness, type 2A
NM_004465.1(FGF10):c.577C>T (p.Arg193Ter)	FGF10	5	44305045	Congenital absence of salivary gland
NM_004465.1(FGF10):c.413G>A (p.Gly138Glu)	FGF10	5	44310443	Congenital absence of salivary gland
NM_000497.3(CYP11B1):c.124C>T (p.Pro42Ser)	CYP11B1	8	142879690	Congenital adrenal hyperplasia;Deficiency of steroid 11-beta-monooxygenase
NM_000497.3(CYP11B1):c.281C>T (p.Pro94Leu)	CYP11B1	8	142879146	Congenital adrenal hyperplasia;Deficiency of steroid 11-beta-monooxygenase
NM_000475.4(NR0B1):c.847C>T (p.Gln283Ter)	NR0B1	X	30308517	Congenital adrenal hypoplasia, X-linked
NM_000475.4(NR0B1):c.1107G>A (p.Trp369Ter)	NR0B1	X	30308257	Congenital adrenal hypoplasia, X-linked
NM_000475.4(NR0B1):c.704G>A (p.Trp235Ter)	NR0B1	X	30308660	Congenital adrenal hypoplasia, X-linked
NM_000475.4(NR0B1):c.513G>A (p.Trp171Ter)	NR0B1	X	30308851	Congenital adrenal hypoplasia, X-linked
NM_000475.4(NR0B1):c.109C>T (p.Gln37Ter)	NR0B1	X	30309255	Congenital adrenal hypoplasia, X-linked
NM_000475.4(NR0B1):c.1183C>T (p.Gln395Ter)	NR0B1	X	30304809	Congenital adrenal hypoplasia, X-linked;not provided
NM_005373.2(MPL):c.556C>T (p.Gln186Ter)	MPL	1	43339435	Congenital amegakaryocytic thrombocytopenia
NM_005373.2(MPL):c.769C>T (p.Arg257Cys)	MPL	1	43340042	Congenital amegakaryocytic thrombocytopenia
NM_005373.2(MPL):c.1904C>T (p.Pro635Leu)	MPL	1	43352768	Congenital amegakaryocytic thrombocytopenia
NM_005373.2(MPL):c.1473G>A (p.Trp491Ter)	MPL	1	43349267	Congenital amegakaryocytic thrombocytopenia
NM_005373.2(MPL):c.127C>T (p.Arg43Ter)	MPL	1	43338146	Congenital amegakaryocytic thrombocytopenia
NM_015375.2(DSTYK):c.654+1G>A	DSTYK	1	205187417	Congenital anomalies of kidney and urinary tract 1, susceptibility to
NM_015375.2(DSTYK):c.24G>A (p.Trp8Ter)	DSTYK	1	205211512	Congenital anomalies of kidney and urinary tract 1, susceptibility to
NM_015375.2(DSTYK):c.655-3C>T	DSTYK	1	205169835	Congenital anomalies of kidney and urinary tract 1, susceptibility to
NM_015375.2(DSTYK):c.86G>A (p.Arg29Gln)	DSTYK	1	205211450	Congenital anomalies of kidney and urinary tract 1, susceptibility to

NM_000492.3(CFTR):c.3208C>T (p.Arg1070Trp)	CFTR	7	117611649	Congenital bilateral absence of the vas deferens;Cystic fibrosis
NM_000492.3(CFTR):c.3209G>A (p.Arg1070Gln)	CFTR	7	117611650	Congenital bilateral absence of the vas deferens;Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.350G>A (p.Arg117His)	CFTR	7	117530975	Congenital bilateral absence of the vas deferens;Cystic fibrosis;Hereditary pancreatitis;ivacaftor response - efficacy;not provided
NM_012064.3(MIP):c.97C>T (p.Arg33Cys)	MIP	12	56454517	Congenital cataract
NM_021954.3(GJA3):c.260C>T (p.Thr87Met)	GJA3	13	20143029	Congenital cataract
NM_021954.3(GJA3):c.176C>T (p.Pro59Leu)	GJA3	13	20143113	Congenital cataract
NM_004715.4(CTDP1):c.863+389C>T	CTDP1	18	79710825	Congenital Cataracts, Facial Dysmorphism, and Neuropathy
NM_170731.4(BDNF):c.29C>T (p.Thr10Ile)	BDNF	11	27658560	Congenital central hypoventilation
NM_001999.3(FBN2):c.3758G>A (p.Cys1253Tyr)	FBN2	5	128335544	Congenital contractural arachnodactyly
NM_001999.3(FBN2):c.1171G>A (p.Glu391Lys)	FBN2	5	128395182	Congenital contractural arachnodactyly
NM_000540.2(RYR1):c.7111G>A (p.Glu2371Lys)	RYR1	19	38499718	Congenital contracture;Delayed gross motor development;Proximal amyotrophy;Short stature
NM_052867.3(NALCN):c.1768C>T (p.Leu590Phe)	NALCN	13	101176371	Congenital contractures of the limbs and face, hypotonia, and developmental delay
NM_052867.3(NALCN):c.979G>A (p.Glu327Lys)	NALCN	13	101292058	Congenital contractures of the limbs and face, hypotonia, and developmental delay
NM_052867.3(NALCN):c.3542G>A (p.Arg1181Gln)	NALCN	13	101083752	Congenital contractures of the limbs and face, hypotonia, and developmental delay;Intellectual disability with episodic ataxia and congenital arthrogryposis
NM_002743.3(PRKCSH):c.1240C>T (p.Gln414Ter)	PRKCSH	19	11448604	Congenital cystic disease of liver
NM_007214.4(SEC63):c.452+1G>A	SEC63	6	107921796	Congenital cystic disease of liver
NM_080669.5(SLC46A1):c.1082-1G>A	SLC46A1	17	28402322	Congenital defect of folate absorption
NM_080669.5(SLC46A1):c.1126C>T (p.Arg376Trp)	SLC46A1	17	28402277	Congenital defect of folate absorption;not provided
NM_080669.5(SLC46A1):c.337C>T (p.Arg113Cys)	SLC46A1	17	28405360	Congenital defect of folate absorption;not provided
NM_080669.5(SLC46A1):c.1127G>A (p.Arg376Gln)	SLC46A1	17	28402276	Congenital defect of folate absorption;not provided
NM_001145293.1(NGLY1):c.1570C>T (p.Arg524Ter)	NGLY1	3	25720179	Congenital disorder of deglycosylation
NM_002435.2(MPI):c.305C>T (p.Ser102Leu)	MPI	15	74891539	Congenital disorder of glycosylation type 1B
NM_002435.2(MPI):c.884G>A (p.Arg295His)	MPI	15	74897050	Congenital disorder of glycosylation type 1B
NM_002435.2(MPI):c.1253G>A (p.Arg418His)	MPI	15	74897711	Congenital disorder of glycosylation type 1B
NM_013339.3(ALG6):c.998C>T (p.Ala333Val)	ALG6	1	63419380	Congenital disorder of glycosylation type 1C
NM_013339.3(ALG6):c.257+5G>A	ALG6	1	63402348	Congenital disorder of glycosylation type 1C;not provided
NM_005787.5(ALG3):c.353G>A (p.Gly118Asp)	ALG3	3	184245559	Congenital disorder of glycosylation type 1D
NM_005787.5(ALG3):c.165C>T (p.Gly55=)	ALG3	3	184248776	Congenital disorder of glycosylation type 1D
NM_005787.5(ALG3):c.512G>A (p.Arg171Gln)	ALG3	3	184245291	Congenital disorder of glycosylation type 1D
NM_024105.3(ALG12):c.200C>T (p.Thr67Met)	ALG12	22	49913480	Congenital disorder of glycosylation type 1G
NM_024105.3(ALG12):c.437G>A (p.Arg146Gln)	ALG12	22	49910466	Congenital disorder of glycosylation type 1G
NM_024105.3(ALG12):c.301G>A (p.Gly101Arg)	ALG12	22	49910602	Congenital disorder of glycosylation type 1G
NM_024079.4(ALG8):c.824G>A (p.Gly275Asp)	ALG8	11	78112724	Congenital disorder of glycosylation type 1H
NM_001382.3(DPAGT1):c.161+5G>A	DPAGT1	11	119101490	Congenital disorder of glycosylation type 1J
NM_019109.4(ALG1):c.434G>A (p.Gly145Asp)	ALG1	16	5075431	Congenital disorder of glycosylation type 1K
NM_019109.4(ALG1):c.826C>T (p.Arg276Trp)	ALG1	16	5078842	Congenital disorder of glycosylation type 1K
NM_019109.4(ALG1):c.773C>T (p.Ser258Leu)	ALG1	16	5078789	Congenital disorder of glycosylation type 1K;Inborn genetic diseases;not provided
NM_052859.3(RFT1):c.199C>T (p.Arg67Cys)	RFT1	3	53123791	Congenital disorder of glycosylation type 1N
NM_052859.3(RFT1):c.892G>A (p.Glu298Lys)	RFT1	3	53105738	Congenital disorder of glycosylation type 1N
NM_052859.3(RFT1):c.1325G>A (p.Arg442Gln)	RFT1	3	53092502	Congenital disorder of glycosylation type 1N
NM_001004127.2(ALG11):c.1192G>A (p.Glu398Lys)	ALG11	13	52024922	Congenital disorder of glycosylation type 1P
NM_024592.4(SRD5A3):c.320G>A (p.Trp107Ter)	SRD5A3	4	55359444	Congenital disorder of glycosylation type 1Q
NM_024592.4(SRD5A3):c.424C>T (p.Arg142Ter)	SRD5A3	4	55364133	Congenital disorder of glycosylation type 1Q

NM_024592.4(SRD5A3):c.603G>A (p.Trp201Ter)	SRD5A3	4	55367628	Congenital disorder of glycosylation type 1Q
NM_024592.4(SRD5A3):c.57G>A (p.Trp19Ter)	SRD5A3	4	55346393	Congenital disorder of glycosylation type 1Q;not provided
NM_002633.2(PGM1):c.1507C>T (p.Arg503Ter)	PGM1	1	63654374	Congenital disorder of glycosylation type 1t
NM_001204527.1(SSR4):c.441+1G>A	SSR4	X	153798137	Congenital disorder of glycosylation type 1y
NM_001204527.1(SSR4):c.442-1G>A	SSR4	X	153798328	Congenital disorder of glycosylation type 1y
NM_004341.4(CAD):c.1843-1G>A	CAD	2	27226130	Congenital disorder of glycosylation type 1z
NM_004341.4(CAD):c.1843-3C>T	CAD	2	27226128	Congenital disorder of glycosylation type 1z
NM_004341.4(CAD):c.5365C>T (p.Arg1789Ter)	CAD	2	27239442	Congenital disorder of glycosylation type 1z
NM_006302.2(MOGS):c.370C>T (p.Gln124Ter)	MOGS	2	74464705	Congenital disorder of glycosylation type 2B
NM_018389.4(SLC35C1):c.439C>T (p.Arg147Cys)	SLC35C1	11	45806240	Congenital disorder of glycosylation type 2C
NM_015386.2(COG4):c.2197C>T (p.Arg733Trp)	COG4	16	70481397	Congenital disorder of glycosylation type 2J
NM_018475.4(TMEM165):c.792+1G>A	TMEM165	4	55418167	Congenital disorder of glycosylation type 2k
NM_018475.4(TMEM165):c.377G>A (p.Arg126His)	TMEM165	4	55411783	Congenital disorder of glycosylation type 2k
NM_018475.4(TMEM165):c.376C>T (p.Arg126Cys)	TMEM165	4	55411782	Congenital disorder of glycosylation type 2k
NM_018475.4(TMEM165):c.910G>A (p.Gly304Arg)	TMEM165	4	55425387	Congenital disorder of glycosylation type 2k
NM_138459.3(NUS1):c.869G>A (p.Arg290His)	NUS1	6	117707002	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE iaa
NM_001042498.2(SLC35A2):c.991G>A (p.Val331Ile)	SLC35A2	X	48904918	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE iim
NM_001042498.2(SLC35A2):c.3G>A (p.Met1Ile)	SLC35A2	X	48911634	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE iim
NM_001042498.2(SLC35A2):c.638C>T (p.Ser213Phe)	SLC35A2	X	48905271	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE iim
NM_152464.2(TMEM199):c.376-1G>A	TMEM199	17	28360528	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE iip;Congenital disorders of glycosylation type II
NM_024740.2(ALG9):c.1588G>A (p.Glu530Lys)	ALG9	11	111836179	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE ii
NM_002435.2(MPI):c.656G>A (p.Arg219Gln)	MPI	15	74893306	Congenital disorder of glycosylation;Congenital disorder of glycosylation type 1B
NM_138477.2(CDAN1):c.3124C>T (p.Arg1042Trp)	CDAN1	15	42726390	Congenital dyserythropoietic anemia, type i
NM_138477.2(CDAN1):c.3389C>T (p.Pro1130Leu)	CDAN1	15	42725550	Congenital dyserythropoietic anemia, type i
NM_138477.2(CDAN1):c.1860+5G>A	CDAN1	15	42731206	Congenital dyserythropoietic anemia, type i
NM_138477.2(CDAN1):c.2140C>T (p.Arg714Trp)	CDAN1	15	42730632	Congenital dyserythropoietic anemia, type i
NM_138477.2(CDAN1):c.2015C>T (p.P672L)	CDAN1	15	42730757	Congenital dyserythropoietic anemia, type i;not provided
NM_032985.4(SEC23B):c.1588C>T (p.Arg530Trp)	SEC23B	20	18543095	Congenital dyserythropoietic anemia, type ii
NM_032985.4(SEC23B):c.790C>T (p.Arg264Ter)	SEC23B	20	18525888	Congenital dyserythropoietic anemia, type ii
NM_032985.4(SEC23B):c.970C>T (p.Arg324Ter)	SEC23B	20	18526508	Congenital dyserythropoietic anemia, type ii
NM_032985.4(SEC23B):c.325G>A (p.Glu109Lys)	SEC23B	20	18515695	Congenital dyserythropoietic anemia, type ii;not provided
NM_006363.4(SEC23B):c.40C>T (p.Arg14Trp)	SEC23B	20	18510875	Congenital dyserythropoietic anemia, type ii;not provided
NM_032985.4(SEC23B):c.649C>T (p.Arg217Ter)	SEC23B	20	18524980	Congenital dyserythropoietic anemia, type ii;not provided
NM_006563.4(KLF1):c.973G>A (p.Glu325Lys)	KLF1	19	12885001	Congenital dyserythropoietic anemia, type IV
NM_057179.2(TWIST2):c.355C>T (p.Gln119Ter)	TWIST2	2	238848570	Congenital ectodermal dysplasia of face
NM_057179.2(TWIST2):c.193C>T (p.Gln65Ter)	TWIST2	2	238848408	Congenital ectodermal dysplasia of face
NM_000375.2(UROS):c.158C>T (p.Pro53Leu)	UROS	10	125815120	Congenital erythropoietic porphyria
NM_000375.2(UROS):c.197C>T (p.Ala66Val)	UROS	10	125815081	Congenital erythropoietic porphyria
NM_000375.2(UROS):c.683C>T (p.Thr228Met)	UROS	10	125788983	Congenital erythropoietic porphyria
NM_000375.2(UROS):c.10C>T (p.Leu4Phe)	UROS	10	125816490	Congenital erythropoietic porphyria
NM_000375.2(UROS):c.562G>A (p.Gly188Arg)	UROS	10	125794978	Congenital erythropoietic porphyria
NM_000375.2(UROS):c.-26-183G>A	UROS	10	125816708	Congenital erythropoietic porphyria
NM_000375.2(UROS):c.673G>A (p.Gly225Ser)	UROS	10	125788993	Congenital erythropoietic porphyria
NM_000375.2(UROS):c.1_63del	UROS	10	125816436	Congenital erythropoietic porphyria
NM_000375.2(UROS):c.311C>T (p.Ala104Val)	UROS	10	125812222	Congenital erythropoietic porphyria

NM_006412.3(AGPAT2):c.202C>T (p.Arg68Ter)	AGPAT2	9	136677537	Congenital generalized lipodystrophy type 1
NM_006412.3(AGPAT2):c.406G>A (p.Gly136Arg)	AGPAT2	9	136677047	Congenital generalized lipodystrophy type 1
NM_006412.3(AGPAT2):c.676C>T (p.Gln226Ter)	AGPAT2	9	136673913	Congenital generalized lipodystrophy type 1
NM_006412.3(AGPAT2):c.514G>A (p.Glu172Lys)	AGPAT2	9	136676659	Congenital generalized lipodystrophy type 1
NM_006412.3(AGPAT2):c.492+1G>A	AGPAT2	9	136676960	Congenital generalized lipodystrophy type 1
NM_006412.3(AGPAT2):c.299G>A (p.Ser100Asn)	AGPAT2	9	136677440	Congenital generalized lipodystrophy type 1
NM_006412.3(AGPAT2):c.194G>A (p.Trp65Ter)	AGPAT2	9	136677545	Congenital generalized lipodystrophy type 1
NM_006412.3(AGPAT2):c.182+1G>A	AGPAT2	9	136687175	Congenital generalized lipodystrophy type 1
NM_006412.3(AGPAT2):c.503G>A (p.Trp168Ter)	AGPAT2	9	136676670	Congenital generalized lipodystrophy type 1
NM_032667.6(BSCL2):c.412C>T (p.Arg138Ter)	BSCL2	11	62694594	Congenital generalized lipodystrophy type 2
NM_032667.6(BSCL2):c.671+5G>A	BSCL2	11	62692371	Congenital generalized lipodystrophy type 2
NM_032667.6(BSCL2):c.823C>T (p.Arg275Ter)	BSCL2	11	62691132	Congenital generalized lipodystrophy type 2
NM_032667.6(BSCL2):c.142C>T (p.Leu48Phe)	BSCL2	11	62705371	Congenital generalized lipodystrophy type 2
NM_032667.6(BSCL2):c.793C>T (p.Arg265Ter)	BSCL2	11	62691300	Congenital generalized lipodystrophy type 2;Encephalopathy, progressive, with or without lipodystrophy
NM_000343.3(SLC5A1):c.82G>A (p.Asp28Asn)	SLC5A1	22	32043363	Congenital glucose-galactose malabsorption
NM_000343.3(SLC5A1):c.1673G>A (p.Arg558His)	SLC5A1	22	32104793	Congenital glucose-galactose malabsorption
NM_001330069.1(PRKD1):c.1798G>A (p.Gly600Arg)	PRKD1	14	29626508	CONGENITAL HEART DEFECTS AND ECTODERMAL DYSPLASIA
NM_031267.3(CDK13):c.2149G>A (p.Gly717Arg)	CDK13	7	39999467	CONGENITAL HEART DEFECTS, DYSMORPHIC FACIAL FEATURES, AND INTELLECTUAL DEVELOPMENTAL DISORDER
NM_031267.3(CDK13):c.2252G>A (p.Arg751Gln)	CDK13	7	40001930	CONGENITAL HEART DEFECTS, DYSMORPHIC FACIAL FEATURES, AND INTELLECTUAL DEVELOPMENTAL DISORDER
NM_015093.5(TAB2):c.622C>T (p.Pro208Ser)	TAB2	6	149378537	Congenital heart defects, multiple types, 2
NM_001145155.1(NR2F2):c.571+1G>A	NR2F2	15	96334604	Congenital heart defects, multiple types, 4
NM_004387.3(NKX2-5):c.391G>A (p.Glu131Lys)	NKX2-5	5	173233153	Congenital heart disease
NM_002052.4(GATA4):c.25G>A (p.Ala9Thr)	GATA4	8	11708337	Congenital heart disease
NM_002052.4(GATA4):c.1263C>T (p.Ser421=)	GATA4	8	11758409	Congenital heart disease
NM_001875.4(CPS1):c.1631C>T (p.Thr544Met)	CPS1	2	210600636	Congenital hyperammonemia, type I
NM_001875.4(CPS1):c.130C>T (p.Gln44Ter)	CPS1	2	210573301	Congenital hyperammonemia, type I
NM_001875.4(CPS1):c.2945G>A (p.Gly982Asp)	CPS1	2	210640045	Congenital hyperammonemia, type I
NM_001875.4(CPS1):c.2359C>T (p.Arg787Ter)	CPS1	2	210608527	Congenital hyperammonemia, type I
NM_000547.5(TPO):c.2395G>A (p.Glu799Lys)	TPO	2	1503956	Congenital hypothyroidism;Deficiency of iodide peroxidase
NM_001171155.1(PET100):c.142C>T (p.Gln48Ter)	PET100	19	7631476	Congenital lactic acidosis;Cytochrome-c oxidase deficiency
NM_000218.2(KCNQ1):c.1766G>A (p.Gly589Asp)	KCNQ1	11	2778009	Congenital long QT syndrome;Jervell and Lange-Nielsen syndrome 1;Long QT syndrome 1;not provided
NM_000218.2(KCNQ1):c.1760C>T (p.Thr587Met)	KCNQ1	11	2778003	Congenital long QT syndrome;Jervell and Lange-Nielsen syndrome 1;Long QT syndrome;not provided
NM_000218.2(KCNQ1):c.604G>A (p.Asp202Asn)	KCNQ1	11	2570754	Congenital long QT syndrome;Jervell and Lange-Nielsen syndrome 1;not provided
NM_000219.5(KCNE1):c.20C>T (p.Thr7Ile)	KCNE1	21	34449615	Congenital long QT syndrome;Jervell and Lange-Nielsen syndrome 2
NM_000218.2(KCNQ1):c.691C>T (p.Arg231Cys)	KCNQ1	11	2572020	Congenital long QT syndrome;KCNQ1-related acquired long QT syndrome;Long QT syndrome 1;not provided
NM_000218.2(KCNQ1):c.905C>T (p.Ala302Val)	KCNQ1	11	2572970	Congenital long QT syndrome;Long QT syndrome
NM_000218.2(KCNQ1):c.815G>A (p.Gly272Asp)	KCNQ1	11	2572880	Congenital long QT syndrome;Long QT syndrome
NM_000238.3(KCNH2):c.2467C>T (p.Arg823Trp)	KCNH2	7	150948981	Congenital long QT syndrome;Long QT syndrome
NM_000218.2(KCNQ1):c.1573G>A (p.Ala525Thr)	KCNQ1	11	2768902	Congenital long QT syndrome;Long QT syndrome 1, recessive;Long QT syndrome, LQT1 subtype;not provided

NM_000238.3(KCNH2):c.2842C>T (p.Arg948Cys)	KCNH2	7	150947729	Congenital long QT syndrome;Long QT syndrome 1/2, digenic	
NM_000218.2(KCNQ1):c.1034G>A (p.Gly345Glu)	KCNQ1	11	2585213	Congenital long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype	
NM_000218.2(KCNQ1):c.350C>T (p.Pro117Leu)	KCNQ1	11	2445448	Congenital long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype	
NM_000218.2(KCNQ1):c.565G>A (p.Gly189Arg)	KCNQ1	11	2570715	Congenital long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.916G>A (p.Gly306Arg)	KCNQ1	11	2572981	Congenital long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.935C>T (p.Thr312Ile)	KCNQ1	11	2583448	Congenital long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.817C>T (p.Leu273Phe)	KCNQ1	11	2572882	Congenital long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.940G>A (p.Gly314Ser)	KCNQ1	11	2583453	Congenital long QT syndrome;Long QT syndrome 1;Long QT syndrome, LQT1 subtype;not provided	
NM_000238.3(KCNH2):c.2453C>T (p.Ser818Leu)	KCNH2	7	150948995	Congenital long QT syndrome;Long QT syndrome 2	
NM_000238.3(KCNH2):c.1882G>A (p.Gly628Ser)	KCNH2	7	150951511	Congenital long QT syndrome;Long QT syndrome 2;not provided	
NM_000238.3(KCNH2):c.1744C>T (p.Arg582Cys)	KCNH2	7	150951649	Congenital long QT syndrome;Long QT syndrome 2;not provided	
NM_000335.4(SCN5A):c.4865G>A (p.Arg1622Gln)	SCN5A	3	38551504	Congenital long QT syndrome;Long QT syndrome 3	
NM_000238.3(KCNH2):c.1468G>A (p.Ala490Thr)	KCNH2	7	150952514	Congenital long QT syndrome;Long QT syndrome, bradycardia-induced;not provided	
NM_000218.2(KCNQ1):c.1024C>T (p.Leu342Phe)	KCNQ1	11	2583537	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.1031C>T (p.Ala344Val)	KCNQ1	11	2583544	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.1876G>A (p.Gly626Ser)	KCNQ1	11	2847848	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.365G>A (p.Cys122Tyr)	KCNQ1	11	2445463	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.409C>T (p.Leu137Phe)	KCNQ1	11	2527950	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.643G>A (p.Val215Met)	KCNQ1	11	2571363	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.794C>T (p.Thr265Ile)	KCNQ1	11	2572859	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.946G>A (p.Gly316Arg)	KCNQ1	11	2583459	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.949G>A (p.Asp317Asn)	KCNQ1	11	2583462	Congenital long QT syndrome;Long QT syndrome, LQT1 subtype;not provided	
NM_000218.2(KCNQ1):c.569G>A (p.Arg190Gln)	KCNQ1	11	2570719	Congenital long QT syndrome;Long QT syndrome 1;not provided	
NM_000218.2(KCNQ1):c.760G>A (p.Val254Met)	KCNQ1	11	2572089	Congenital long QT syndrome;Long QT syndrome 1;not provided	
NM_000218.2(KCNQ1):c.1663C>T (p.Arg555Cys)	KCNQ1	11	2776032	Congenital long QT syndrome;Long QT syndrome 1;not provided	
NM_000218.2(KCNQ1):c.805G>A (p.Gly269Ser)	KCNQ1	11	2572870	Congenital long QT syndrome;Long QT syndrome 1;not provided	
NM_000218.2(KCNQ1):c.806G>A (p.Gly269Asp)	KCNQ1	11	2572871	Congenital long QT syndrome;Long QT syndrome 1;not provided	
NM_000218.2(KCNQ1):c.1637C>T (p.Ser546Leu)	KCNQ1	11	2776006	Congenital long QT syndrome;Long QT syndrome 1;not provided	
NM_000218.2(KCNQ1):c.502G>A (p.Gly168Arg)	KCNQ1	11	2570652	Congenital long QT syndrome;Long QT syndrome 1;not provided	
NM_000238.3(KCNH2):c.1801G>A (p.Gly601Ser)	KCNH2	7	150951592	Congenital long QT syndrome;Long QT syndrome 2	
NM_000238.3(KCNH2):c.1682C>T (p.Ala561Val)	KCNH2	7	150951711	Congenital long QT syndrome;Long QT syndrome 2;not provided	
NM_000238.3(KCNH2):c.1841C>T (p.Ala614Val)	KCNH2	7	150951552	Congenital long QT syndrome;Long QT syndrome 2;not provided	
NM_000238.3(KCNH2):c.1750G>A (p.Gly584Ser)	KCNH2	7	150951643	Congenital long QT syndrome;Long QT syndrome 2;Short QT syndrome 1;not provided	
NM_000218.2(KCNQ1):c.1096C>T (p.Arg366Trp)	KCNQ1	11	2585275	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.1615C>T (p.Arg539Trp)	KCNQ1	11	2775984	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.1664G>A (p.Arg555His)	KCNQ1	11	2776033	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.520C>T (p.Arg174Cys)	KCNQ1	11	2570670	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.674C>T (p.Ser225Leu)	KCNQ1	11	2571394	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.724G>A (p.Asp242Asn)	KCNQ1	11	2572053	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.727C>T (p.Arg243Cys)	KCNQ1	11	2572056	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.775C>T (p.Arg259Cys)	KCNQ1	11	2572104	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.776G>A (p.Arg259His)	KCNQ1	11	2572105	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.830C>T (p.Ser277Leu)	KCNQ1	11	2572895	Congenital long QT syndrome;Long QT syndrome;not provided	
NM_000218.2(KCNQ1):c.973G>A (p.Gly325Arg)	KCNQ1	11	2583486	Congenital long QT syndrome;Long QT syndrome;not provided	

NM_000238.3(KCNH2):c.1501G>A (p.Asp501Asn)	KCNH2	7	150952481	Congenital long QT syndrome;Long QT syndrome;not provided
NM_000238.3(KCNH2):c.1600C>T (p.Arg534Cys)	KCNH2	7	150951793	Congenital long QT syndrome;Long QT syndrome;not provided
NM_000238.3(KCNH2):c.1810G>A (p.Gly604Ser)	KCNH2	7	150951583	Congenital long QT syndrome;Long QT syndrome;not provided
NM_000719.6(CACNA1C):c.1216G>A (p.Gly406Arg)	CACNA1C	12	2504944	Congenital long QT syndrome;Long QT syndrome;Timothy syndrome;not provided
NM_000218.2(KCNQ1):c.575G>A (p.Arg192His)	KCNQ1	11	2570725	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.1001C>T (p.Pro334Leu)	KCNH2	7	150957418	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.1264G>A (p.Ala422Thr)	KCNH2	7	150952718	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.1421C>T (p.Thr474Ile)	KCNH2	7	150952561	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.164C>T (p.Ser55Leu)	KCNH2	7	150974854	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.1681G>A (p.Ala561Thr)	KCNH2	7	150951712	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.1714G>A (p.Gly572Ser)	KCNH2	7	150951679	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.1825G>A (p.Asp609Asn)	KCNH2	7	150951568	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.1894C>T (p.Pro632Ser)	KCNH2	7	150951499	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.215C>T (p.Pro72Leu)	KCNH2	7	150974803	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.2254C>T (p.Arg752Trp)	KCNH2	7	150950312	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.2417G>A (p.Gly806Glu)	KCNH2	7	150949031	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.2458G>A (p.Gly820Arg)	KCNH2	7	150948990	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.2477C>T (p.Thr826Ile)	KCNH2	7	150948971	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.2680C>T (p.Arg894Cys)	KCNH2	7	150948456	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.2843G>A (p.Arg948His)	KCNH2	7	150947728	Congenital long QT syndrome;not provided
NM_000238.3(KCNH2):c.299G>A (p.Arg100Gln)	KCNH2	7	150974719	Congenital long QT syndrome;not provided
NM_000891.2(KCNJ2):c.200G>A (p.Arg67Gln)	KCNJ2	17	70175239	Congenital long QT syndrome;not provided
NM_000891.2(KCNJ2):c.224C>T (p.Thr75Met)	KCNJ2	17	70175263	Congenital long QT syndrome;not provided
NM_198056.2(SCN5A):c.3691G>A (p.Glu1231Lys)	SCN5A	3	38566558	Congenital long QT syndrome;not provided
NM_198056.2(SCN5A):c.3883G>A (p.Glu1295Lys)	SCN5A	3	38562495	Congenital long QT syndrome;not provided
NM_198056.2(SCN5A):c.3988G>A (p.Ala1330Thr)	SCN5A	3	38560404	Congenital long QT syndrome;not provided
NM_198056.2(SCN5A):c.4934C>T (p.Thr1645Met)	SCN5A	3	38551438	Congenital long QT syndrome;not provided
NM_198056.2(SCN5A):c.5287G>A (p.Val1763Met)	SCN5A	3	38551085	Congenital long QT syndrome;not provided
NM_000719.6(CACNA1C):c.1204G>A (p.Gly402Ser)	CACNA1C	12	2504932	Congenital long QT syndrome;Timothy syndrome;not provided
NM_001080467.2(MYO5B):c.1125G>A (p.Trp375Ter)	MYO5B	18	49974547	Congenital microvillous atrophy
NM_001080467.2(MYO5B):c.1966C>T (p.Arg656Cys)	MYO5B	18	49936289	Congenital microvillous atrophy
NM_001080467.2(MYO5B):c.1979C>T (p.Pro660Leu)	MYO5B	18	49936276	Congenital microvillous atrophy
NM_000426.3(LAMA2):c.4645C>T (p.Arg1549Ter)	LAMA2	6	129353285	Congenital muscular dystrophy due to partial LAMA2 deficiency
NM_000426.3(LAMA2):c.7147C>T (p.Arg2383Ter)	LAMA2	6	129464444	Congenital muscular dystrophy due to partial LAMA2 deficiency;Merosin deficient congenital muscular dystrophy;not provided
NM_170707.3(LMNA):c.428C>T (p.Ser143Phe)	LMNA	1	156130688	Congenital muscular dystrophy, LMNA-related;not provided
NM_024301.4(FKRP):c.1343C>T (p.Pro448Leu)	FKRP	19	46756793	Congenital muscular dystrophy-dystroglycanopathy (with or without mental retardation) type B5;Congenital muscular dystrophy-dystroglycanopathy without mental retardation, type B5
NM_024301.4(FKRP):c.953G>A (p.Cys318Tyr)	FKRP	19	46756403	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A5
NM_013382.5(POMT2):c.2177G>A (p.Gly726Glu)	POMT2	14	77277452	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A2;Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B2
NM_013382.5(POMT2):c.1057G>A (p.Gly353Ser)	POMT2	14	77296223	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A2;Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B2
NM_013382.5(POMT2):c.1912C>T (p.Arg638Ter)	POMT2	14	77278849	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A2;Limb-girdle muscular dystrophy-dystroglycanopathy, type C2

NM_013382.5(POMT2):c.1006+1G>A	POMT2	14	77298688	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A2;Limb-girdle muscular dystrophy-dystroglycanopathy, type C2
NM_004737.5(LARGE1):c.992C>T (p.Ser331Phe)	LARGE1	22	33384205	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A6
NM_001101426.3(ISPD):c.802C>T (p.Arg268Ter)	ISPD	7	16301454	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A7
NM_001101426.3(ISPD):c.466G>A (p.Asp156Asn)	ISPD	7	16406129	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A7
NM_001101426.3(ISPD):c.643C>T (p.Gln215Ter)	ISPD	7	16376133	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A7
NM_007171.3(POMT1):c.193G>A (p.Gly65Arg)	POMT1	9	131506184	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B1
NM_007171.3(POMT1):c.2005G>A (p.Ala669Thr)	POMT1	9	131522160	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B1;Limb-girdle muscular dystrophy-dystroglycanopathy, type C1;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 1;not provided
NM_007171.3(POMT1):c.1540C>T (p.Arg514Ter)	POMT1	9	131518945	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B1;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 1;not provided
NM_013382.5(POMT2):c.1941G>A (p.Trp647Ter)	POMT2	14	77278820	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B2
NM_013382.5(POMT2):c.737G>A (p.Gly246Asp)	POMT2	14	77301169	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B2
NM_017739.3(POMGNT1):c.1539+1G>A	POMGNT1	1	46192097	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B3;Limb-girdle muscular dystrophy-dystroglycanopathy, type C3;Muscle eye brain disease;POMGNT1-Related Disorders;not provided
NM_017739.3(POMGNT1):c.652+1G>A	POMGNT1	1	46194843	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B3;Muscle eye brain disease
NM_017739.3(POMGNT1):c.1469G>A (p.Cys490Tyr)	POMGNT1	1	46192168	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B3;Muscle eye brain disease
NM_017739.3(POMGNT1):c.643C>T (p.Arg215Ter)	POMGNT1	1	46194853	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B3;Muscle eye brain disease
NM_004737.5(LARGE1):c.1525G>A (p.Glu509Lys)	LARGE1	22	33304434	Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B6
NM_006306.3(SMC1A):c.1487G>A (p.Arg496His)	SMC1A	X	53409120	Congenital muscular hypertrophy-cerebral syndrome
NM_006306.3(SMC1A):c.2368C>T (p.Arg790Trp)	SMC1A	X	53403618	Congenital muscular hypertrophy-cerebral syndrome
NM_006306.3(SMC1A):c.2131C>T (p.Arg711Trp)	SMC1A	X	53405077	Congenital muscular hypertrophy-cerebral syndrome
NM_006306.3(SMC1A):c.1193G>A (p.Arg398Gln)	SMC1A	X	53411822	Congenital muscular hypertrophy-cerebral syndrome
NM_006306.3(SMC1A):c.586C>T (p.Arg196Cys)	SMC1A	X	53413261	Congenital muscular hypertrophy-cerebral syndrome
NM_006306.3(SMC1A):c.547C>T (p.Gln183Ter)	SMC1A	X	53413300	Congenital muscular hypertrophy-cerebral syndrome
NM_006306.3(SMC1A):c.2369G>A (p.Arg790Gln)	SMC1A	X	53403617	Congenital muscular hypertrophy-cerebral syndrome
NM_006306.3(SMC1A):c.3146G>A (p.Arg1049Gln)	SMC1A	X	53382645	Congenital muscular hypertrophy-cerebral syndrome;not provided
NM_198576.3(AGRN):c.226G>A (p.Gly76Ser)	AGRN	1	1022225	Congenital myasthenic syndrome
NM_198576.3(AGRN):c.5023G>A (p.Gly1675Ser)	AGRN	1	1050473	Congenital myasthenic syndrome
NM_198576.3(AGRN):c.5611G>A (p.Gly1871Arg)	AGRN	1	1051775	Congenital myasthenic syndrome
NM_000334.4(SCN4A):c.4360C>T (p.Arg1454Trp)	SCN4A	17	63941922	Congenital myasthenic syndrome
NM_001382.3(DPAGT1):c.349G>A (p.Val117Ile)	DPAGT1	11	119100777	Congenital myasthenic syndrome 13
NM_001039523.2(CHRNA1):c.988G>A (p.Val330Ile)	CHRNA1	2	174750035	Congenital myasthenic syndrome 1B, fast-channel
NM_001244710.1(GFPT1):c.331C>T (p.Arg111Cys)	GFPT1	2	69363563	Congenital myasthenic syndrome with tubular aggregates 1
NM_003172.3(SURF1):c.751C>T (p.Gln251Ter)	SURF1	9	133352446	Congenital myasthenic syndrome, acetazolamide-responsive
NM_003172.3(SURF1):c.371G>A (p.Gly124Glu)	SURF1	9	133353893	Congenital myasthenic syndrome, acetazolamide-responsive
NM_000334.4(SCN4A):c.4370G>A (p.Arg1457His)	SCN4A	17	63941912	Congenital myasthenic syndrome, acetazolamide-responsive
NM_198576.3(AGRN):c.1057C>T (p.Gln353Ter)	AGRN	1	1041582	Congenital myasthenic syndrome;Myasthenic syndrome, congenital, 8
NM_001100.3(ACTA1):c.1000C>T (p.Pro334Ser)	ACTA1	1	229431633	Congenital myopathy with fiber type disproportion
NM_001100.3(ACTA1):c.143G>A (p.Gly48Asp)	ACTA1	1	229432867	Congenital myopathy with fiber type disproportion

NM_001100.3(ACTA1):c.16G>A (p.Glu6Lys)	ACTA1	1	229433100	Congenital myopathy with fiber type disproportion
NM_001100.3(ACTA1):c.727G>A (p.Glu243Lys)	ACTA1	1	229432075	Congenital myopathy with fiber type disproportion
NM_020451.2(SELENON):c.943G>A (p.Gly315Ser)	SELENON	1	25809753	Congenital myopathy with fiber type disproportion;Eichsfeld type congenital muscular dystrophy;not provided
NM_152263.3(TPM3):c.11C>T (p.Ala4Val)	TPM3	1	154192008	Congenital myopathy with fiber type disproportion;not provided
NM_152263.3(TPM3):c.721G>A (p.Glu241Lys)	TPM3	1	154170454	Congenital myopathy with fiber type disproportion;not provided
NM_000083.2(CLCN1):c.566C>T (p.Ser189Phe)	CLCN1	7	143321718	Congenital myotonia, autosomal dominant form
NM_000083.2(CLCN1):c.950G>A (p.Arg317Gln)	CLCN1	7	143330868	Congenital myotonia, autosomal dominant form;Congenital myotonia, autosomal recessive form;Myotonia congenita
NM_000083.2(CLCN1):c.1439C>T (p.Pro480Leu)	CLCN1	7	143339290	Congenital myotonia, autosomal dominant form;Myotonia congenita
NM_000083.2(CLCN1):c.689G>A (p.Gly230Glu)	CLCN1	7	143321841	Congenital myotonia, autosomal dominant form;Myotonia congenita;not provided
NM_000083.2(CLCN1):c.871G>A (p.Glu291Lys)	CLCN1	7	143330789	Congenital myotonia, autosomal recessive form
NM_000083.2(CLCN1):c.1495G>A (p.Gly499Arg)	CLCN1	7	143339534	Congenital myotonia, autosomal recessive form
NM_001130145.2(YAP1):c.370C>T (p.Arg124Ter)	YAP1	11	102114192	Congenital ocular coloboma
NM_198897.1(FIBP):c.673C>T (p.Gln225Ter)	FIBP	11	65885181	Congenital ocular coloboma;Large hands;Macrocephaly;Overgrowth;THAUVIN-ROBINET-FAIVRE SYNDROME;facial dysmorphism;learning disabilities
NM_005216.4(DDOST):c.650G>A (p.Gly217Asp)	DDOST	1	20654660	Congenital order of glycosylation type 1r
NM_000111.2(SLC26A3):c.1386G>A (p.Trp462Ter)	SLC26A3	7	107779689	Congenital secretory diarrhea, chloride type
NM_206933.2(USH2A):c.11864G>A (p.Trp3955Ter)	USH2A	1	215728232	Congenital sensorineural hearing impairment;Hearing impairment;Retinitis pigmentosa 39;Usher syndrome, type 2A;not provided
NM_002506.2(NGF):c.661C>T (p.Arg221Trp)	NGF	1	115286135	Congenital sensory neuropathy with selective loss of small myelinated fibers
NM_024769.3(CLMP):c.821G>A (p.Arg274Gln)	CLMP	11	123074702	Congenital short bowel syndrome
NM_024769.3(CLMP):c.664C>T (p.Arg222Ter)	CLMP	11	123083100	Congenital short bowel syndrome
NM_024769.3(CLMP):c.508C>T (p.Arg170Ter)	CLMP	11	123083728	Congenital short bowel syndrome
NM_000539.3(RHO):c.269G>A (p.Gly90Asp)	RHO	3	129529002	Congenital stationary night blindness, autosomal dominant 1
NM_000539.3(RHO):c.281C>T (p.Thr94Ile)	RHO	3	129529014	Congenital stationary night blindness, autosomal dominant 1
NM_000539.3(RHO):c.491C>T (p.Ala164Val)	RHO	3	129531005	Congenital stationary night blindness, autosomal dominant 1
NM_144499.2(GNAT1):c.113G>A (p.Gly38Asp)	GNAT1	3	50193139	Congenital stationary night blindness, autosomal dominant 3
NM_022567.2(NYX):c.1049G>A (p.Trp350Ter)	NYX	X	41474502	Congenital stationary night blindness, type 1A;not provided
NM_000843.3(GRM6):c.1462C>T (p.Gln488Ter)	GRM6	5	178986876	Congenital stationary night blindness, type 1B
NM_000843.3(GRM6):c.1861C>T (p.Arg621Ter)	GRM6	5	178986393	Congenital stationary night blindness, type 1B;not provided
NM_000843.3(GRM6):c.2341G>A (p.Glu781Lys)	GRM6	5	178983005	Congenital stationary night blindness, type 1B;not provided
NM_000843.3(GRM6):c.2122C>T (p.Gln708Ter)	GRM6	5	178986132	Congenital stationary night blindness, type 1B;not provided
NM_000843.3(GRM6):c.448G>A (p.Gly150Ser)	GRM6	5	178994497	Congenital stationary night blindness, type 1B;not provided
NM_000843.3(GRM6):c.137C>T (p.Pro46Leu)	GRM6	5	178994808	Congenital stationary night blindness, type 1B;not provided
NM_000843.3(GRM6):c.1565G>A (p.Cys522Tyr)	GRM6	5	178986689	Congenital stationary night blindness, type 1B;not provided
NM_002420.5(TRPM1):c.31C>T (p.Gln11Ter)	TRPM1	15	31070213	Congenital stationary night blindness, type 1C
NM_002420.5(TRPM1):c.1870C>T (p.Arg624Cys)	TRPM1	15	31042102	Congenital stationary night blindness, type 1C
NM_002420.5(TRPM1):c.2998C>T (p.Arg1000Ter)	TRPM1	15	31031046	Congenital stationary night blindness, type 1C;not provided
NM_001004334.3(GPR179):c.598C>T (p.Arg200Ter)	GPR179	17	38343192	Congenital stationary night blindness, type 1E
NM_001004334.3(GPR179):c.1784+1G>A	GPR179	17	38334703	Congenital stationary night blindness, type 1E
NM_001004334.3(GPR179):c.673C>T (p.Gln225Ter)	GPR179	17	38343117	Congenital stationary night blindness, type 1E
NM_001004334.3(GPR179):c.1807C>T (p.His603Tyr)	GPR179	17	38334016	Congenital stationary night blindness, type 1E;not provided
NM_198506.4(LRIT3):c.983G>A (p.Cys328Tyr)	LRIT3	4	109869732	Congenital stationary night blindness, type 1F
NM_198506.4(LRIT3):c.1318C>T (p.Arg440Ter)	LRIT3	4	109870067	Congenital stationary night blindness, type 1F

NM_005183.3(CACNA1F):c.1166G>A (p.Gly369Asp)	CACNA1F	X	49228048	Congenital stationary night blindness, type 2A
NM_005183.3(CACNA1F):c.2905C>T (p.Arg969Ter)	CACNA1F	X	49218511	Congenital stationary night blindness, type 2A
NM_005183.3(CACNA1F):c.2683C>T (p.Arg895Ter)	CACNA1F	X	49219344	Congenital stationary night blindness, type 2A
NM_005183.3(CACNA1F):c.2576+1G>A	CACNA1F	X	49219633	Congenital stationary night blindness, type 2A
NM_145200.3(CABP4):c.370C>T (p.Arg124Cys)	CABP4	11	67456191	Congenital stationary night blindness, type 2B
NM_145200.3(CABP4):c.646C>T (p.Arg216Ter)	CABP4	11	67457677	Congenital stationary night blindness, type 2B
NM_000097.5(CPOX):c.991C>T (p.Arg331Trp)	CPOX	3	98585622	Coproporphyria
NM_000097.5(CPOX):c.85C>T (p.Gln29Ter)	CPOX	3	98593420	Coproporphyria
NM_000097.5(CPOX):c.623C>T (p.Ser208Phe)	CPOX	3	98591089	Coproporphyria
NM_000097.5(CPOX):c.982C>T (p.Arg328Cys)	CPOX	3	98585631	Coproporphyria
NM_000097.5(CPOX):c.1339C>T (p.Arg447Cys)	CPOX	3	98580709	Coproporphyria;Hereditary coproporphyria
NM_007035.3(KERA):c.520C>T (p.Gln174Ter)	KERA	12	91055762	Cornea plana 2
NM_007035.3(KERA):c.937C>T (p.Arg313Ter)	KERA	12	91051468	Cornea plana 2
NM_007035.3(KERA):c.835C>T (p.Arg279Ter)	KERA	12	91055447	Cornea plana 2
NM_001174089.1(SLC4A11):c.1415G>A (p.Arg472Lys)	SLC4A11	20	3230515	Corneal dystrophy and perceptive deafness
NM_001174089.1(SLC4A11):c.2078G>A (p.Gly693Glu)	SLC4A11	20	3228952	Corneal dystrophy, Fuchs endothelial, 4
NM_001174089.1(SLC4A11):c.2213C>T (p.Thr738Met)	SLC4A11	20	3228687	Corneal dystrophy, Fuchs endothelial, 4
NM_001174089.1(SLC4A11):c.1147G>A (p.Glu383Lys)	SLC4A11	20	3230954	Corneal dystrophy, Fuchs endothelial, 4
NM_152336.3(AGBL1):c.3220C>T (p.Arg1074Ter)	AGBL1	15	86674435	Corneal dystrophy, Fuchs endothelial, 8
NM_001174089.1(SLC4A11):c.2216G>A (p.Arg739Gln)	SLC4A11	20	3228684	Corneal endothelial dystrophy
NM_001174089.1(SLC4A11):c.1418C>T (p.Ser473Leu)	SLC4A11	20	3230258	Corneal endothelial dystrophy
NM_001174089.1(SLC4A11):c.1343G>A (p.Gly448Asp)	SLC4A11	20	3230587	Corneal endothelial dystrophy
NM_001174089.1(SLC4A11):c.1765C>T (p.Arg589Ter)	SLC4A11	20	3229430	Corneal endothelial dystrophy
NM_001174089.1(SLC4A11):c.2557C>T (p.Arg853Cys)	SLC4A11	20	3228260	Corneal endothelial dystrophy
NM_001174089.1(SLC4A11):c.2558G>A (p.Arg853His)	SLC4A11	20	3228259	Corneal endothelial dystrophy
NM_001127464.2(ZNF469):c.10016G>A (p.Cys3339Tyr)	ZNF469	16	88437570	Corneal fragility keratoglobus, blue sclerae AND joint hypermobility
NM_001127464.2(ZNF469):c.9184C>T (p.Arg3062Ter)	ZNF469	16	88436738	Corneal fragility keratoglobus, blue sclerae AND joint hypermobility
NM_001033053.2(NLRP1):c.160G>A (p.Ala54Thr)	NLRP1	17	5583798	Corneal intraepithelial dyskeratosis and ectodermal dysplasia
NM_033004.3(NLRP1):c.197C>T (p.Ala66Val)	NLRP1	17	5583761	Corneal intraepithelial dyskeratosis and ectodermal dysplasia;not provided
NM_012293.2(PXDN):c.2638C>T (p.Arg880Cys)	PXDN	2	1649142	Corneal opacification with other ocular anomalies
NM_012293.2(PXDN):c.1021C>T (p.Arg341Ter)	PXDN	2	1666484	Corneal opacification with other ocular anomalies
NM_133433.3(NIPBL):c.5167C>T (p.Arg1723Ter)	NIPBL	5	37020615	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.4606C>T (p.Arg1536Ter)	NIPBL	5	37014728	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.133C>T (p.Arg45Ter)	NIPBL	5	36955540	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.3445C>T (p.Arg1149Ter)	NIPBL	5	37000513	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.7327C>T (p.Gln2443Ter)	NIPBL	5	37057249	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.64+1G>A	NIPBL	5	36953761	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.598C>T (p.Gln200Ter)	NIPBL	5	36962262	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.610+1G>A	NIPBL	5	36962275	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.771+1G>A	NIPBL	5	36971037	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.772-1G>A	NIPBL	5	36971944	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.869-1G>A	NIPBL	5	36975775	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.892C>T (p.Gln298Ter)	NIPBL	5	36975799	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.922C>T (p.Arg308Ter)	NIPBL	5	36975829	Cornelia de Lange syndrome 1
NM_133433.3(NIPBL):c.1183C>T (p.Gln395Ter)	NIPBL	5	36976090	Cornelia de Lange syndrome 1

NM_133433.3(NIPBL):c.1297C>T (p.Gln433Ter)	NIPBL	5	36976204	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.1372C>T (p.Gln458Ter)	NIPBL	5	36976279	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.1576C>T (p.Gln526Ter)	NIPBL	5	36984756	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.1885C>T (p.Arg629Ter)	NIPBL	5	36985065	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.2260C>T (p.Arg754Ter)	NIPBL	5	36985440	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.2389C>T (p.Arg797Ter)	NIPBL	5	36985569	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.2422C>T (p.Arg808Ter)	NIPBL	5	36985602	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.2494C>T (p.Arg832Ter)	NIPBL	5	36985674	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.2500C>T (p.Arg834Ter)	NIPBL	5	36985680	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.4320+1G>A	NIPBL	5	37008089	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5328+1G>A	NIPBL	5	37020878	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5335C>T (p.Arg1779Ter)	NIPBL	5	37022057	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5365C>T (p.Arg1789Ter)	NIPBL	5	37022087	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5455C>T (p.Arg1819Ter)	NIPBL	5	37022271	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5482C>T (p.Arg1828Ter)	NIPBL	5	37022298	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5483G>A (p.Arg1828Gln)	NIPBL	5	37022299	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5709+1G>A	NIPBL	5	37024720	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5808+1G>A	NIPBL	5	37026328	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5862+1G>A	NIPBL	5	37027413	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.6565C>T (p.Gln2189Ter)	NIPBL	5	37046175	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.6590G>A (p.Gly2197Glu)	NIPBL	5	37048502	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.6892C>T (p.Arg2298Cys)	NIPBL	5	37049239	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.6893G>A (p.Arg2298His)	NIPBL	5	37049240	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.6952C>T (p.Gln2318Ter)	NIPBL	5	37049299	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.6954+1G>A	NIPBL	5	37049302	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.7102C>T (p.Gln2368Ter)	NIPBL	5	37052405	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.7168G>A (p.Ala2390Thr)	NIPBL	5	37052471	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.7849C>T (p.Gln2617Ter)	NIPBL	5	37061007	Cornelia de Lange syndrome 1	
NM_005933.3(KMT2A):c.2233C>T (p.Arg745Ter)	KMT2A	11	118473392	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.2521C>T (p.Arg841Ter)	NIPBL	5	36985701	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.4495C>T (p.Gln1499Ter)	NIPBL	5	37010160	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.5044C>T (p.Arg1682Ter)	NIPBL	5	37020492	Cornelia de Lange syndrome 1	
NM_015384.4(NIPBL):c.6082C>T (p.Gln2028Ter)	NIPBL	5	37038712	Cornelia de Lange syndrome 1	
NM_015384.4(NIPBL):c.7150C>T (p.Gln2384Ter)	NIPBL	5	37052453	Cornelia de Lange syndrome 1	
NM_133433.3(NIPBL):c.2602C>T (p.Arg868Ter)	NIPBL	5	36985782	Cornelia de Lange syndrome 1;not provided	
NM_015384.4(NIPBL):c.5440C>T (p.Arg1814Ter)	NIPBL	5	37022256	Cornelia de Lange syndrome 1;not provided	
NM_133433.3(NIPBL):c.7219C>T (p.Arg2407Ter)	NIPBL	5	37052522	Cornelia de Lange syndrome 1;not provided	
NM_133433.3(NIPBL):c.8377C>T (p.Arg2793Ter)	NIPBL	5	37064854	Cornelia de Lange syndrome 1;not provided	
NM_005445.3(SMC3):c.1964G>A (p.Gly655Asp)	SMC3	10	110596398	Cornelia de Lange syndrome 3	
NM_005445.3(SMC3):c.1462G>A (p.Glu488Lys)	SMC3	10	110589944	Cornelia de Lange syndrome 3	
NM_018486.2(HDAC8):c.164+5G>A	HDAC8	X	72572052	Cornelia de Lange syndrome 5	
NM_018486.2(HDAC8):c.932C>T (p.Thr311Met)	HDAC8	X	72462077	Cornelia de Lange syndrome 5	
NM_001166419.1(HDAC8):c.356C>T (p.Thr119Met)	HDAC8	X	72567970	Cornelia de Lange syndrome 5	
NM_018486.2(HDAC8):c.229C>T (p.Gln77Ter)	HDAC8	X	72568820	Cornelia de Lange syndrome 5	
NM_018486.2(HDAC8):c.490C>T (p.Arg164Ter)	HDAC8	X	72495216	Cornelia de Lange syndrome 5;not provided	

NM_002336.2(LRP6):c.1831C>T (p.Arg611Cys)	LRP6	12	12164494	Coronary artery disease, autosomal dominant 2
NM_002336.2(LRP6):c.1418G>A (p.Arg473Gln)	LRP6	12	12179937	Coronary artery disease, autosomal dominant 2
NM_002336.2(LRP6):c.1079G>A (p.Arg360His)	LRP6	12	12181337	Coronary artery disease, autosomal dominant 2
NM_005544.2(IRS1):c.2911G>A (p.Gly971Arg)	IRS1	2	226795828	Coronary artery disease, susceptibility to;Insulin resistance, susceptibility to
NM_000040.1(APOC3):c.55+1G>A	APOC3	11	116830638	Coronary heart disease;Hyperalphalipoproteinemia 2
NM_000040.1(APOC3):c.127G>A (p.Ala43Thr)	APOC3	11	116830844	Coronary heart disease;Hyperalphalipoproteinemia 2
NM_000040.1(APOC3):c.55C>T (p.Arg19Ter)	APOC3	11	116830637	Coronary heart disease;Hyperalphalipoproteinemia 2;not provided
NM_005215.3(DCC):c.2260G>A (p.Val754Met)	DCC	18	53339808	Corpus callosum agenesis
NM_005215.3(DCC):c.2414G>A (p.Gly805Glu)	DCC	18	53386097	Corpus callosum agenesis
NM_005215.3(DCC):c.2677G>A (p.Ala893Thr)	DCC	18	53391876	Corpus callosum agenesis
NM_005215.3(DCC):c.823C>T (p.Arg275Ter)	DCC	18	52923832	Corpus callosum agenesis;Mirror movements 1
NM_000425.4(L1CAM):c.719C>T (p.Pro240Leu)	L1CAM	X	153870475	Corpus callosum, partial agenesis of, X-linked;X-linked hydrocephalus syndrome
NM_001197181.1(TUBB3):c.397G>A (p.Glu133Lys)	TUBB3	16	89935064	Cortical dysplasia, complex, with other brain malformations 1
NM_001197181.1(TUBB3):c.689C>T (p.Ala230Val)	TUBB3	16	89935356	Cortical dysplasia, complex, with other brain malformations 1
NM_006086.3(TUBB3):c.1228G>A (p.Glu410Lys)	TUBB3	16	89935679	Cortical dysplasia, complex, with other brain malformations 1;Fibrosis of extraocular muscles, congenital, 3a, with or without extraocular involvement
NM_006086.3(TUBB3):c.533C>T (p.Thr178Met)	TUBB3	16	89934984	Cortical dysplasia, complex, with other brain malformations 1;not provided
NM_004522.2(KIF5C):c.709G>A (p.Glu237Lys)	KIF5C	2	148947018	Cortical dysplasia, complex, with other brain malformations 2;not provided
NM_001098511.2(KIF2A):c.950G>A (p.Ser317Asn)	KIF2A	5	62361319	Cortical dysplasia, complex, with other brain malformations 3
NM_178014.3(TUBB):c.1057G>A (p.Val353Ile)	TUBB	6	30724119	Cortical dysplasia, complex, with other brain malformations 6
NM_178014.3(TUBB):c.1201G>A (p.Glu401Lys)	TUBB	6	30724263	Cortical dysplasia, complex, with other brain malformations 6
NM_178014.3(TUBB):c.662C>T (p.Thr221Ile)	TUBB	6	30723724	Cortical dysplasia, complex, with other brain malformations 6
NM_006059.3(LAMC3):c.1156C>T (p.Gln386Ter)	LAMC3	9	131039043	Cortical malformations, occipital
NM_001756.3(SERPINA6):c.1165G>A (p.Asp389Asn)	SERPINA6	14	94304471	Corticosteroid-binding globulin deficiency
NM_000498.3(CYP11B2):c.554C>T (p.Thr185Ile)	CYP11B2	8	142915087	Corticosterone methyloxidase type 2 deficiency
NM_000498.3(CYP11B2):c.814C>T (p.Gln272Ter)	CYP11B2	8	142914404	Corticosterone methyloxidase type 2 deficiency
NM_004285.3(H6PD):c.960G>A (p.Val320=)	H6PD	1	9262273	Cortisone reductase deficiency 1
NM_004285.3(H6PD):c.1076G>A (p.Gly359Asp)	H6PD	1	9263569	Cortisone reductase deficiency 1
NM_181755.2(HSD11B1):c.409C>T (p.Arg137Cys)	HSD11B1	1	209707020	Cortisone reductase deficiency 2
NM_176795.4(HRAS):c.173C>T (p.Thr58Ile)	HRAS	11	533883	Costello syndrome
NM_005343.3(HRAS):c.35G>A (p.Gly12Asp)	HRAS	11	534288	Costello syndrome;Costello syndrome, severe;Neoplasm of breast;Nevus sebaceous;Rasopathy;not provided
NM_000314.6(PTEN):c.386G>A (p.Gly129Glu)	PTEN	10	87933145	Cowden syndrome 1;not provided
NM_006218.3(PIK3CA):c.403G>A (p.Glu135Lys)	PIK3CA	3	179199740	Cowden syndrome 5
NM_006218.3(PIK3CA):c.652G>A (p.Glu218Lys)	PIK3CA	3	179201379	Cowden syndrome 5
NM_006218.3(PIK3CA):c.1066G>A (p.Val356Ile)	PIK3CA	3	179204509	Cowden syndrome 5
NM_006218.3(PIK3CA):c.1145G>A (p.Arg382Lys)	PIK3CA	3	179204588	Cowden syndrome 5
NM_001014431.1(AKT1):c.73C>T (p.Arg25Cys)	AKT1	14	104780190	Cowden syndrome 6
NM_025237.2(SOST):c.61G>A (p.Val21Met)	SOST	17	43758681	Craniodiaphyseal dysplasia, autosomal dominant
NM_052985.3(IFT122):c.1118C>T (p.Ser373Phe)	IFT122	3	129476463	Craniectodermal dysplasia 1
NM_052985.3(IFT122):c.502+5G>A	IFT122	3	129461309	Craniectodermal dysplasia 1
NM_052985.3(IFT122):c.1636G>A (p.Gly546Arg)	IFT122	3	129479917	Craniectodermal dysplasia 1
NM_001006657.1(WDR35):c.2623G>A (p.Ala875Thr)	WDR35	2	19933469	Craniectodermal dysplasia 2
NM_001006657.1(WDR35):c.1288+1G>A	WDR35	2	19960553	Craniectodermal dysplasia 2
NM_025132.3(WDR19):c.3307C>T (p.Arg1103Ter)	WDR19	4	39268040	Craniectodermal dysplasia 4

NM_019026.4(TMCO1):c.412C>T (p.Arg138Ter)	TMCO1	1	165752166	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome
NM_005631.4(SMO):c.1234C>T (p.Leu412Phe)	SMO	7	129206557	Craniofacial malformations, asymmetric, with polysyndactyly and abnormal skin and gut development
NM_004429.4(EFNB1):c.332C>T (p.Thr111Ile)	EFNB1	X	68838820	Craniofrontonasal dysplasia
NM_004429.4(EFNB1):c.451G>A (p.Gly151Ser)	EFNB1	X	68839708	Craniofrontonasal dysplasia
NM_004429.4(EFNB1):c.110G>A (p.Trp37Ter)	EFNB1	X	68829886	Craniofrontonasal dysplasia
NM_004429.4(EFNB1):c.496C>T (p.Gln166Ter)	EFNB1	X	68839753	Craniofrontonasal dysplasia
NM_004429.4(EFNB1):c.161C>T (p.Pro54Leu)	EFNB1	X	68838649	Craniofrontonasal dysplasia;not provided
NM_004429.4(EFNB1):c.196C>T (p.Arg66Ter)	EFNB1	X	68838684	Craniofrontonasal dysplasia;not provided
NM_000165.4(GJA1):c.716G>A (p.Arg239Gln)	GJA1	6	121447563	Craniometaphyseal dysplasia, autosomal recessive type
NM_000474.3(TWIST1):c.556G>A (p.Ala186Thr)	TWIST1	7	19116766	Craniosynostosis 1
NM_000474.3(TWIST1):c.563C>T (p.Ser188Leu)	TWIST1	7	19116759	Craniosynostosis 1
NM_002449.4(MSX2):c.443C>T (p.Pro148Leu)	MSX2	5	174729222	Craniosynostosis 2
NM_207036.1(TCF12):c.1838G>A (p.Arg613His)	TCF12	15	57273122	Craniosynostosis 3
NM_207036.1(TCF12):c.1876C>T (p.Arg626Ter)	TCF12	15	57273160	Craniosynostosis 3
NM_006494.3(ERF):c.547C>T (p.Arg183Ter)	ERF	19	42249565	Craniosynostosis 4
NM_006494.3(ERF):c.194G>A (p.Arg65Gln)	ERF	19	42250394	Craniosynostosis 4
NM_006494.3(ERF):c.1270C>T (p.Gln424Ter)	ERF	19	42248842	Craniosynostosis 4
NM_006494.3(ERF):c.256C>T (p.Arg86Cys)	ERF	19	42250332	Craniosynostosis 4;not provided
NM_003412.3(ZIC1):c.1165C>T (p.Gln389Ter)	ZIC1	3	147413372	Craniosynostosis 6
NM_001142784.2(IL11RA):c.886C>T (p.Arg296Trp)	IL11RA	9	34659834	Craniosynostosis and dental anomalies
NM_001142784.2(IL11RA):c.475C>T (p.Gln159Ter)	IL11RA	9	34657331	Craniosynostosis and dental anomalies
NM_000141.4(FGFR2):c.1032G>A (p.Ala344=)	FGFR2	10	121517371	Craniosynostosis,Craniosynostosis, nonclassifiable autosomal dominant;Crouzon syndrome;Scaphocephaly and axenfeld-rieger anomaly
NM_021224.5(ZNF462):c.3787C>T (p.Arg1263Ter)	ZNF462	9	106927699	Craniosynostosis;Mental retardation, autosomal dominant
NM_005629.3(SLC6A8):c.1540C>T (p.Arg514Ter)	SLC6A8	X	153694577	Creatine deficiency, X-linked
NM_005629.3(SLC6A8):c.259G>A (p.Gly87Arg)	SLC6A8	X	153688833	Creatine deficiency, X-linked
NM_005629.3(SLC6A8):c.1631C>T (p.Pro544Leu)	SLC6A8	X	153694753	Creatine deficiency, X-linked
NM_005629.3(SLC6A8):c.1661C>T (p.Pro554Leu)	SLC6A8	X	153694783	Creatine deficiency, X-linked
NM_033337.2(CAV3):c.80G>A (p.Arg27Gln)	CAV3	3	8733956	Creatine phosphokinase, elevated serum;Distal myopathy, Tateyama type;Limb-girdle muscular dystrophy, type 1C;Rippling muscle disease;Rippling muscle disease 2;not provided
NM_033337.2(CAV3):c.136G>A (p.Ala46Thr)	CAV3	3	8745547	Creatine phosphokinase, elevated serum;Limb-girdle muscular dystrophy, type 1C;Rippling muscle disease 2;not provided
NM_033337.2(CAV3):c.86C>T (p.Pro29Leu)	CAV3	3	8733962	Creatine phosphokinase, elevated serum;not provided
NM_000463.2(UGT1A1):c.1124C>T (p.Ser375Phe)	UGT1A1	2	233768259	Crigler Najjar syndrome, type 1
NM_000463.2(UGT1A1):c.991C>T (p.Gln331Ter)	UGT1A1	2	233767160	Crigler Najjar syndrome, type 1
NM_000463.2(UGT1A1):c.923G>A (p.Gly308Glu)	UGT1A1	2	233767092	Crigler Najjar syndrome, type 1
NM_000463.2(UGT1A1):c.145C>T (p.Gln49Ter)	UGT1A1	2	233760432	Crigler Najjar syndrome, type 1
NM_000463.2(UGT1A1):c.1021C>T (p.Arg341Ter)	UGT1A1	2	233767873	Crigler Najjar syndrome, type 1;Gilbert's syndrome
NM_000463.2(UGT1A1):c.1069C>T (p.Gln357Ter)	UGT1A1	2	233767921	Crigler-Najjar syndrome
NM_000141.4(FGFR2):c.1013G>A (p.Gly338Glu)	FGFR2	10	121517390	Crouzon syndrome
NM_000141.4(FGFR2):c.1025G>A (p.Cys342Tyr)	FGFR2	10	121517378	Crouzon syndrome;Pfeiffer syndrome
NM_207361.5(FREM2):c.5920G>A (p.Glu1974Lys)	FREM2	13	38784709	Cryptophthalmos syndrome
NM_207361.5(FREM2):c.5914G>A (p.Glu1972Lys)	FREM2	13	38784703	Cryptophthalmos syndrome
NM_025074.6(FRAS1):c.8602C>T (p.Gln2868Ter)	FRAS1	4	78481962	Cryptophthalmos syndrome
NM_025074.6(FRAS1):c.9013C>T (p.Gln3005Ter)	FRAS1	4	78496859	Cryptophthalmos syndrome

NM_025074.6(FRAS1):c.3799C>T (p.Gln1267Ter)	FRAS1	4	78387525	Cryptophthalmos syndrome
NM_025074.6(FRAS1):c.370C>T (p.Arg124Ter)	FRAS1	4	78252452	Cryptophthalmos syndrome
NM_025074.6(FRAS1):c.2722+1G>A	FRAS1	4	78364055	Cryptophthalmos syndrome;not provided
NM_005543.3(INSL3):c.278C>T (p.Pro93Leu)	INSL3	19	17816972	Cryptorchidism, unilateral or bilateral
NM_005543.3(INSL3):c.304C>T (p.Arg102Cys)	INSL3	19	17816946	Cryptorchidism, unilateral or bilateral
NM_005543.3(INSL3):c.305G>A (p.Arg102His)	INSL3	19	17816945	Cryptorchidism, unilateral or bilateral
NM_005543.3(INSL3):c.217C>T (p.Arg73Ter)	INSL3	19	17817033	Cryptorchidism, unilateral or bilateral
NM_005515.3(MNX1):c.775C>T (p.Gln259Ter)	MNX1	7	157006556	Currarino triad
NM_005450.4(NOG):c.668C>T (p.Pro223Leu)	NOG	17	56594891	Cushing's symphalangism
NM_005450.4(NOG):c.551G>A (p.Cys184Tyr)	NOG	17	56594774	Cushing's symphalangism
NM_000455.4(STK11):c.508C>T (p.Gln170Ter)	STK11	19	1220416	Cutaneous malignant melanoma 1;Neoplasm
NM_001100119.1(XRCC3):c.722C>T (p.Thr241Met)	XRCC3	14	103699416	Cutaneous malignant melanoma 6
NM_012463.3(ATP6V0A2):c.2293C>T (p.Gln765Ter)	ATP6V0A2	12	123754537	Cutis laxa with osteodystrophy
NM_012463.3(ATP6V0A2):c.294+1G>A	ATP6V0A2	12	123722449	Cutis laxa with osteodystrophy
NM_012463.3(ATP6V0A2):c.304C>T (p.Gln102Ter)	ATP6V0A2	12	123724663	Cutis laxa with osteodystrophy
NM_012463.3(ATP6V0A2):c.187C>T (p.Arg63Ter)	ATP6V0A2	12	123718692	Cutis laxa with osteodystrophy;not provided
NM_012463.3(ATP6V0A2):c.1514+1G>A	ATP6V0A2	12	123744785	Cutis laxa with osteodystrophy;not provided
NM_001042544.1(LTBP4):c.1453C>T (p.Arg485Ter)	LTBP4	19	40608315	Cutis laxa with severe pulmonary, gastrointestinal, and urinary abnormalities
NM_002860.3(ALDH18A1):c.413G>A (p.Arg138Gln)	ALDH18A1	10	95637327	Cutis laxa, autosomal dominant 3
NM_002860.3(ALDH18A1):c.412C>T (p.Arg138Trp)	ALDH18A1	10	95637328	Cutis laxa, autosomal dominant 3;not provided
NM_001696.3(ATP6V1E1):c.634C>T (p.Arg212Trp)	ATP6V1E1	22	17592721	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IIC
NM_001690.3(ATP6V1A):c.1012C>T (p.Arg338Cys)	ATP6V1A	3	113794895	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IID
NM_001690.3(ATP6V1A):c.215G>A (p.Gly72Asp)	ATP6V1A	3	113784227	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IID
NM_000052.6(ATP7A):c.1910C>T (p.Ser637Leu)	ATP7A	X	78011216	Cutis laxa, X-linked;Menkes kinky-hair syndrome
NM_001171.5(ABCC6):c.3421C>T (p.Arg1141Ter)	ABCC6	16	16163078	Cutis laxa;Generalized arterial calcification of infancy 2;Papule;Pseudoxanthoma elasticum;Pseudoxanthoma elasticum, forme fruste;not provided
NM_002860.3(ALDH18A1):c.251G>A (p.Arg84Gln)	ALDH18A1	10	95643044	Cutis laxa-corneal clouding-oligophrenia syndrome
NM_002860.3(ALDH18A1):c.2350C>T (p.His784Tyr)	ALDH18A1	10	95606800	Cutis laxa-corneal clouding-oligophrenia syndrome
NM_002860.3(ALDH18A1):c.1923+1G>A	ALDH18A1	10	95613741	Cutis laxa-corneal clouding-oligophrenia syndrome
NM_000184.2(HBG2):c.190C>T (p.His64Tyr)	HBG2	11	5254417	Cyanosis, transient neonatal
NM_000184.2(HBG2):c.277C>T (p.His93Tyr)	HBG2	11	5254330	Cyanosis, transient neonatal
NM_000184.2(HBG2):c.202G>A (p.Val68Met)	HBG2	11	5254405	Cyanosis, transient neonatal
NM_001972.3(ELANE):c.659G>A (p.Arg220Gln)	ELANE	19	856019	Cyclical neutropenia
NM_001972.3(ELANE):c.182C>T (p.Ala61Val)	ELANE	19	852990	Cyclical neutropenia
NM_001972.3(ELANE):c.598-1G>A	ELANE	19	855957	Cyclical neutropenia
NM_015247.2(CYLD):c.1327C>T (p.Gln443Ter)	CYLD	16	50779853	Cylindromatosis, familial
NM_015247.2(CYLD):c.1363C>T (p.Gln455Ter)	CYLD	16	50779889	Cylindromatosis, familial
NM_015247.2(CYLD):c.2569C>T (p.Gln857Ter)	CYLD	16	50794311	Cylindromatosis, familial
NM_015247.2(CYLD):c.2806C>T (p.Arg936Ter)	CYLD	16	50796443	Cylindromatosis, familial;Familial multiple trichoepithelioma;Spiegler-Brooke syndrome
NM_015247.2(CYLD):c.2272C>T (p.Arg758Ter)	CYLD	16	50792627	Cylindromatosis, familial;not specified
NM_001902.5(CTH):c.200C>T (p.Thr67Ile)	CTH	1	70415987	Cystathioninuria
NM_000492.3(CFTR):c.1477C>T (p.Gln493Ter)	CFTR	7	117559548	Cystic fibrosis
NM_000492.3(CFTR):c.2538G>A (p.Trp846Ter)	CFTR	7	117594977	Cystic fibrosis
NM_000492.3(CFTR):c.3947G>A (p.Trp1316Ter)	CFTR	7	117652915	Cystic fibrosis
NM_000492.3(CFTR):c.3484C>T (p.Arg1162Ter)	CFTR	7	117627537	Cystic fibrosis

NM_000492.3(CFTR):c.2551C>T (p.Arg851Ter)	CFTR	7	117594990	Cystic fibrosis
NM_000492.3(CFTR):c.3472C>T (p.Arg1158Ter)	CFTR	7	117627525	Cystic fibrosis
NM_000492.3(CFTR):c.1475C>T (p.Ser492Phe)	CFTR	7	117559546	Cystic fibrosis
NM_000492.3(CFTR):c.1679G>A (p.Arg560Lys)	CFTR	7	117587833	Cystic fibrosis
NM_000492.3(CFTR):c.3197G>A (p.Arg1066His)	CFTR	7	117611638	Cystic fibrosis
NM_000492.3(CFTR):c.3873+1G>A	CFTR	7	117642594	Cystic fibrosis
NM_000492.3(CFTR):c.3196C>T (p.Arg1066Cys)	CFTR	7	117611637	Cystic fibrosis
NM_000492.3(CFTR):c.1766+1G>A	CFTR	7	117590440	Cystic fibrosis
NM_000492.3(CFTR):c.1046C>T (p.Ala349Val)	CFTR	7	117540276	Cystic fibrosis
NM_000492.3(CFTR):c.2490+1G>A	CFTR	7	117592658	Cystic fibrosis
NM_000492.3(CFTR):c.3712C>T (p.Gln1238Ter)	CFTR	7	117627765	Cystic fibrosis
NM_000492.3(CFTR):c.3718-1G>A	CFTR	7	117642437	Cystic fibrosis
NM_000492.3(CFTR):c.171G>A (p.Trp57Ter)	CFTR	7	117509040	Cystic fibrosis
NM_000492.3(CFTR):c.3937C>T (p.Gln1313Ter)	CFTR	7	117652905	Cystic fibrosis
NM_000492.3(CFTR):c.274G>A (p.Glu92Lys)	CFTR	7	117530899	Cystic fibrosis
NM_000492.3(CFTR):c.271G>A (p.Gly91Arg)	CFTR	7	117509140	Cystic fibrosis
NM_000492.3(CFTR):c.1013C>T (p.Thr338Ile)	CFTR	7	117540243	Cystic fibrosis
NM_000492.3(CFTR):c.3266G>A (p.Trp1089Ter)	CFTR	7	117611707	Cystic fibrosis
NM_000492.3(CFTR):c.1055G>A (p.Arg352Gln)	CFTR	7	117540285	Cystic fibrosis
NM_000492.3(CFTR):c.1654C>T (p.Gln552Ter)	CFTR	7	117587808	Cystic fibrosis
NM_000492.3(CFTR):c.2668C>T (p.Gln890Ter)	CFTR	7	117603542	Cystic fibrosis
NM_000492.3(CFTR):c.2845C>T (p.His949Tyr)	CFTR	7	117603719	Cystic fibrosis
NM_000492.3(CFTR):c.3611G>A (p.Trp1204Ter)	CFTR	7	117627664	Cystic fibrosis
NM_000492.3(CFTR):c.3659C>T (p.Thr1220Ile)	CFTR	7	117627712	Cystic fibrosis
NM_000492.3(CFTR):c.3746G>A (p.Gly1249Glu)	CFTR	7	117642466	Cystic fibrosis
NM_000492.3(CFTR):c.1658G>A (p.Arg553Gln)	CFTR	7	117587812	Cystic fibrosis
NM_000492.3(CFTR):c.1585-8G>A	CFTR	7	117587731	Cystic fibrosis
NM_000492.3(CFTR):c.223C>T (p.Arg75Ter)	CFTR	7	117509092	Cystic fibrosis
NM_000492.3(CFTR):c.1680-1G>A	CFTR	7	117590352	Cystic fibrosis
NM_000492.3(CFTR):c.349C>T (p.Arg117Cys)	CFTR	7	117530974	Cystic fibrosis
NM_000492.3(CFTR):c.1203G>A (p.Trp401Ter)	CFTR	7	117542102	Cystic fibrosis
NM_000492.3(CFTR):c.1240C>T (p.Gln414Ter)	CFTR	7	117548671	Cystic fibrosis
NM_000492.3(CFTR):c.1202G>A (p.Trp401Ter)	CFTR	7	117542101	Cystic fibrosis
NM_000492.3(CFTR):c.1209+1G>A	CFTR	7	117542109	Cystic fibrosis
NM_000492.3(CFTR):c.115C>T (p.Gln39Ter)	CFTR	7	117504314	Cystic fibrosis
NM_000492.3(CFTR):c.1116+1G>A	CFTR	7	117540347	Cystic fibrosis
NM_000492.3(CFTR):c.1393-1G>A	CFTR	7	117559463	Cystic fibrosis
NM_000492.3(CFTR):c.1573C>T (p.Gln525Ter)	CFTR	7	117559644	Cystic fibrosis
NM_000492.3(CFTR):c.164+1G>A	CFTR	7	117504364	Cystic fibrosis
NM_000492.3(CFTR):c.166G>A (p.Glu56Lys)	CFTR	7	117509035	Cystic fibrosis
NM_000492.3(CFTR):c.170G>A (p.Trp57Ter)	CFTR	7	117509039	Cystic fibrosis
NM_000492.3(CFTR):c.2053C>T (p.Gln685Ter)	CFTR	7	117592220	Cystic fibrosis
NM_000492.3(CFTR):c.2125C>T (p.Arg709Ter)	CFTR	7	117592292	Cystic fibrosis
NM_000492.3(CFTR):c.2290C>T (p.Arg764Ter)	CFTR	7	117592457	Cystic fibrosis
NM_000492.3(CFTR):c.2353C>T (p.Arg785Ter)	CFTR	7	117592520	Cystic fibrosis

NM_000492.3(CFTR):c.2374C>T (p.Arg792Ter)	CFTR	7	117592541	Cystic fibrosis
NM_000492.3(CFTR):c.2537G>A (p.Trp846Ter)	CFTR	7	117594976	Cystic fibrosis
NM_000492.3(CFTR):c.292C>T (p.Gln98Ter)	CFTR	7	117530917	Cystic fibrosis
NM_000492.3(CFTR):c.2989-1G>A	CFTR	7	117610518	Cystic fibrosis
NM_000492.3(CFTR):c.3293G>A (p.Trp1098Ter)	CFTR	7	117611734	Cystic fibrosis
NM_000492.3(CFTR):c.4144C>T (p.Gln1382Ter)	CFTR	7	117665466	Cystic fibrosis
NM_000492.3(CFTR):c.4231C>T (p.Gln1411Ter)	CFTR	7	117665553	Cystic fibrosis
NM_000492.3(CFTR):c.4234C>T (p.Gln1412Ter)	CFTR	7	117665556	Cystic fibrosis
NM_000492.3(CFTR):c.579+5G>A	CFTR	7	117534370	Cystic fibrosis
NM_000492.3(CFTR):c.595C>T (p.His199Tyr)	CFTR	7	117535263	Cystic fibrosis
NM_000492.3(CFTR):c.613C>T (p.Pro205Ser)	CFTR	7	117535281	Cystic fibrosis
NM_000492.3(CFTR):c.658C>T (p.Gln220Ter)	CFTR	7	117535326	Cystic fibrosis
NM_000492.3(CFTR):c.830G>A (p.Trp277Ter)	CFTR	7	117536634	Cystic fibrosis
NM_000492.3(CFTR):c.1117G>A (p.Asp373Asn)	CFTR	7	117542016	Cystic fibrosis
NM_000492.3(CFTR):c.3963+1G>A	CFTR	7	117652932	Cystic fibrosis
NM_000492.3(CFTR):c.1117-1G>A	CFTR	7	117542015	Cystic fibrosis
NM_000492.3(CFTR):c.3294G>A (p.Trp1098Ter)	CFTR	7	117611735	Cystic fibrosis
NM_000492.3(CFTR):c.1657C>T (p.Arg553Ter)	CFTR	7	117587811	Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.1675G>A (p.Ala559Thr)	CFTR	7	117587829	Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.1865G>A (p.Gly622Asp)	CFTR	7	117592032	Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.165-1G>A	CFTR	7	117509033	Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.200C>T (p.Pro67Leu)	CFTR	7	117509069	Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.2834C>T (p.Ser945Leu)	CFTR	7	117603708	Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.743+1G>A	CFTR	7	117535412	Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.1679+1G>A	CFTR	7	117587834	Cystic fibrosis;Hereditary pancreatitis
NM_000492.3(CFTR):c.3846G>A (p.Trp1282Ter)	CFTR	7	117642566	Cystic fibrosis;Hereditary pancreatitis;ataluren response - Efficacy;not provided
NM_000492.3(CFTR):c.1652G>A (p.Gly551Asp)	CFTR	7	117587806	Cystic fibrosis;Hereditary pancreatitis;ivacaftor response - Efficacy;not provided
NM_000492.3:c.3718-2477C>T	CFTR	7	117639961	Cystic fibrosis;Hereditary pancreatitis;not provided
NM_000492.3(CFTR):c.2988+1G>A	CFTR	7	117606754	Cystic fibrosis;Hereditary pancreatitis;not provided
NM_000492.3(CFTR):c.2988G>A (p.Gln996=)	CFTR	7	117606753	Cystic fibrosis;Hereditary pancreatitis;not provided
NM_000492.3(CFTR):c.2657+5G>A	CFTR	7	117602868	Cystic fibrosis;Hereditary pancreatitis;not provided
NM_000492.3(CFTR):c.274-1G>A	CFTR	7	117530898	Cystic fibrosis;Hereditary pancreatitis;not provided
NM_000492.3(CFTR):c.3612G>A (p.Trp1204Ter)	CFTR	7	117627665	Cystic fibrosis;Hereditary pancreatitis;not provided
NM_000492.3(CFTR):c.4426C>T (p.Gln1476Ter)	CFTR	7	117667091	Cystic fibrosis;Hereditary pancreatitis;not provided
NM_000492.3(CFTR):c.1646G>A (p.Ser549Asn)	CFTR	7	117587800	Cystic fibrosis;ivacaftor response - Efficacy
NM_000492.3(CFTR):c.3752G>A (p.Ser1251Asn)	CFTR	7	117642472	Cystic fibrosis;ivacaftor response - Efficacy
NM_000492.3(CFTR):c.4046G>A (p.Gly1349Asp)	CFTR	7	117664770	Cystic fibrosis;ivacaftor response - Efficacy
NM_000492.3(CFTR):c.532G>A (p.Gly178Arg)	CFTR	7	117534318	Cystic fibrosis;ivacaftor response - Efficacy
NM_000492.3(CFTR):c.3731G>A (p.Gly1244Glu)	CFTR	7	117642451	Cystic fibrosis;ivacaftor response - Efficacy
NM_000492.3(CFTR):c.1651G>A (p.Gly551Ser)	CFTR	7	117587805	Cystic fibrosis;ivacaftor response - Efficacy;not provided
NM_000492.3(CFTR):c.1585-1G>A	CFTR	7	117587738	Cystic fibrosis;not provided
NM_000492.3(CFTR):c.1000C>T (p.Arg334Trp)	CFTR	7	117540230	Cystic fibrosis;not provided
NM_000492.3(CFTR):c.254G>A (p.Gly85Glu)	CFTR	7	117509123	Cystic fibrosis;not provided
NM_000492.3(CFTR):c.1040G>A (p.Arg347His)	CFTR	7	117540270	Cystic fibrosis;not provided
NM_000492.3(CFTR):c.273+1G>A	CFTR	7	117509143	Cystic fibrosis;not provided

NM_004937.2(CTNS):c.382C>T (p.Gln128Ter)	CTNS	17	3655273	Cystinosis
NM_004937.2(CTNS):c.922G>A (p.Gly308Arg)	CTNS	17	3659927	Cystinosis
NM_004937.2(CTNS):c.589G>A (p.Gly197Arg)	CTNS	17	3656703	Cystinosis, ocular nonnephropathic;Nephropathic cystinosis
NM_004937.2(CTNS):c.416C>T (p.Ser139Phe)	CTNS	17	3655307	Cystinosis;Cystinosis, atypical nephropathic;Juvenile nephropathic cystinosis;Nephropathic cystinosis
NM_014270.4(SLC7A9):c.508G>A (p.Val170Met)	SLC7A9	19	32862557	Cystinuria
NM_014270.4(SLC7A9):c.313G>A (p.Gly105Arg)	SLC7A9	19	32864261	Cystinuria
NM_014270.4(SLC7A9):c.583G>A (p.Gly195Arg)	SLC7A9	19	32862482	Cystinuria
NM_014270.4(SLC7A9):c.775G>A (p.Gly259Arg)	SLC7A9	19	32859939	Cystinuria
NM_014270.4(SLC7A9):c.997C>T (p.Arg333Trp)	SLC7A9	19	32843932	Cystinuria
NM_014270.4(SLC7A9):c.782C>T (p.Pro261Leu)	SLC7A9	19	32859932	Cystinuria
NM_014270.4(SLC7A9):c.368C>T (p.Thr123Met)	SLC7A9	19	32864206	Cystinuria
NM_000341.3(SLC3A1):c.542G>A (p.Arg181Gln)	SLC3A1	2	44280827	Cystinuria
NM_000341.3(SLC3A1):c.1085G>A (p.Arg362His)	SLC3A1	2	44301076	Cystinuria
NM_000341.3(SLC3A1):c.808C>T (p.Arg270Ter)	SLC3A1	2	44286074	Cystinuria
m.6328C>T	MT-CO1	MT	6328	Cytochrome c oxidase i deficiency
NM_001136194.1(FASTKD2):c.1294C>T (p.Arg432Ter)	FASTKD2	2	206774264	Cytochrome-c oxidase deficiency
NM_004589.3(SCO1):c.521C>T (p.Pro174Leu)	SCO1	17	10692805	Cytochrome-c oxidase deficiency
NM_001303.3(COX10):c.674C>T (p.Pro225Leu)	COX10	17	14159926	Cytochrome-c oxidase deficiency
m.9952G>A	MT-CO3	MT	9952	Cytochrome-c oxidase deficiency
m.9379G>A	MT-CO3	MT	9379	Cytochrome-c oxidase deficiency
m.7896G>A	MT-CO2	MT	7896	Cytochrome-c oxidase deficiency
m.6480G>A	MT-CO1	MT	6480	Cytochrome-c oxidase deficiency
m.6930G>A	MT-CO1	MT	6930	Cytochrome-c oxidase deficiency
NM_001863.4(COX6B1):c.59G>A (p.Arg20His)	COX6B1	19	35651302	Cytochrome-c oxidase deficiency
NM_001257133.1(COX14):c.57G>A (p.Met19Ile)	COX14	12	50120100	Cytochrome-c oxidase deficiency
NM_004589.3(SCO1):c.394G>A (p.Gly132Ser)	SCO1	17	10692932	Cytochrome-c oxidase deficiency
NM_032374.4(APOPT1):c.235C>T (p.Arg79Ter)	APOPT1	14	103571695	Cytochrome-c oxidase deficiency
NM_032374.4(APOPT1):c.163-1G>A	APOPT1	14	103571622	Cytochrome-c oxidase deficiency
NM_001863.4(COX6B1):c.58C>T (p.Arg20Cys)	COX6B1	19	35651301	Cytochrome-c oxidase deficiency
NM_001844.4(COL2A1):c.823C>T (p.Arg275Cys)	COL2A1	12	47994041	Czech dysplasia metatarsal type
NM_152783.4(D2HGDH):c.566C>T (p.Pro189Leu)	D2HGDH	2	241743697	D-2-hydroxyglutaric aciduria 1
NM_001122606.1(LAMP2):c.520C>T (p.Gln174Ter)	LAMP2	X	120449006	Danon disease
NM_002294.2(LAMP2):c.1093+1G>A	LAMP2	X	120441729	Danon disease
NM_002294.2(LAMP2):c.183+1G>A	LAMP2	X	120456650	Danon disease
NM_002294.2(LAMP2):c.294G>A (p.Trp98Ter)	LAMP2	X	120455460	Danon disease
NM_002294.2(LAMP2):c.962G>A (p.Trp321Ter)	LAMP2	X	120441861	Danon disease
NM_002294.2(LAMP2):c.928G>A (p.Val310Ile)	LAMP2	X	120442599	Danon disease;not provided
NM_002294.2(LAMP2):c.293G>A (p.Trp98Ter)	LAMP2	X	120455461	Danon disease;not provided
NM_002294.2(LAMP2):c.877C>T (p.Arg293Ter)	LAMP2	X	120442650	Danon disease;not provided
NM_170665.3(ATP2A2):c.2305G>A (p.Gly769Arg)	ATP2A2	12	110342435	Darier disease, segmental;not provided
NM_032229.2(SLITRK6):c.1240C>T (p.Gln414Ter)	SLITRK6	13	85795269	Deafness and myopia
NM_032229.2(SLITRK6):c.541C>T (p.Arg181Ter)	SLITRK6	13	85795968	Deafness and myopia
NM_000466.2(PEX1):c.3750G>A (p.Trp1250Ter)	PEX1	7	92489310	Deafness enamel hypoplasia nail defects
NM_005247.2(FGF3):c.310C>T (p.Arg104Ter)	FGF3	11	69816334	Deafness with labyrinthine aplasia microtia and microdontia (LAMM)
NM_005247.2(FGF3):c.283C>T (p.Arg95Trp)	FGF3	11	69816361	Deafness with labyrinthine aplasia microtia and microdontia (LAMM);not provided

NM_000260.3(MYO7A):c.652G>A (p.Asp218Asn)	MYO7A	11	77156921	Deafness, autosomal dominant 11;Usher syndrome, type 1
NM_000260.3(MYO7A):c.689C>T (p.Ala230Val)	MYO7A	11	77156958	Deafness, autosomal dominant 11;Usher syndrome, type 1
NM_005422.2(TECTA):c.6062G>A (p.Arg2021His)	TECTA	11	121187894	Deafness, autosomal dominant 12
NM_005422.2(TECTA):c.5668C>T (p.Arg1890Cys)	TECTA	11	121168135	Deafness, autosomal dominant 12
NM_005422.2(TECTA):c.2887G>A (p.Ala963Thr)	TECTA	11	121130157	Deafness, autosomal dominant 12
NM_080680.2(COL11A2):c.3100C>T (p.Arg1034Cys)	COL11A2	6	33171763	Deafness, autosomal dominant 13
NM_080680.2(COL11A2):c.2423G>A (p.Gly808Glu)	COL11A2	6	33174534	Deafness, autosomal dominant 13
NM_002700.2(POU4F3):c.865C>T (p.Leu289Phe)	POU4F3	5	146340292	Deafness, autosomal dominant 15
NM_002700.2(POU4F3):c.977G>A (p.Arg326Lys)	POU4F3	5	146340404	Deafness, autosomal dominant 15
NM_153700.2(STRC):c.4057C>T (p.Gln1353Ter)	STRC	15	43604720	Deafness, autosomal dominant 16;not provided
NM_001614.3(ACTG1):c.266C>T (p.Thr89Ile)	ACTG1	17	81512000	Deafness, autosomal dominant 20
NM_001614.3(ACTG1):c.791C>T (p.Pro264Leu)	ACTG1	17	81511199	Deafness, autosomal dominant 20
NM_001614.3(ACTG1):c.833C>T (p.Thr278Ile)	ACTG1	17	81511078	Deafness, autosomal dominant 20
NM_001614.3(ACTG1):c.721G>A (p.Glu241Lys)	ACTG1	17	81511269	Deafness, autosomal dominant 20;Nonsyndromic hearing loss and deafness;not provided
NM_004999.3(MYO6):c.1325G>A (p.Cys442Tyr)	MYO6	6	75857198	Deafness, autosomal dominant 22
NM_004999.3(MYO6):c.2545C>T (p.Arg849Ter)	MYO6	6	75886881	Deafness, autosomal dominant 22
NM_005982.3(SIX1):c.373G>A (p.Glu125Lys)	SIX1	14	60648817	Deafness, autosomal dominant 23
NM_139319.2(SLC17A8):c.632C>T (p.Ala211Val)	SLC17A8	12	100396373	Deafness, autosomal dominant 25
NM_024915.3(GRHL2):c.1258-1G>A	GRHL2	8	101631636	Deafness, autosomal dominant 28
NM_138691.2(TMC1):c.1714G>A (p.Asp572Asn)	TMC1	9	72816161	Deafness, autosomal dominant 36;Nonsyndromic hearing loss and deafness
NM_004004.5(GJB2):c.535G>A (p.Asp179Asn)	GJB2	13	20189047	Deafness, autosomal dominant 3a
NM_004004.5(GJB2):c.598G>A (p.Gly200Arg)	GJB2	13	20188984	Deafness, autosomal dominant 3a;Deafness, autosomal recessive 1A
NM_004004.5(GJB2):c.71G>A (p.Trp24Ter)	GJB2	13	20189511	Deafness, autosomal dominant 3a;Deafness, autosomal recessive 1A;Hearing impairment;Nonsyndromic hearing loss and deafness;not provided
NM_004004.5(GJB2):c.416G>A (p.Ser139Asn)	GJB2	13	20189166	Deafness, autosomal dominant 3a;Deafness, autosomal recessive 1A;Hearing impairment;Nonsyndromic hearing loss and deafness;not provided
NM_004004.5(GJB2):c.224G>A (p.Arg75Gln)	GJB2	13	20189358	Deafness, autosomal dominant 3a;Deafness, autosomal recessive 1A;Keratoderma palmoplantar deafness;Nonsyndromic hearing loss and deafness;not provided
NM_004004.5(GJB2):c.95G>A (p.Arg32His)	GJB2	13	20189487	Deafness, autosomal dominant 3a;Deafness, autosomal recessive 1A;Nonsyndromic hearing loss and deafness
NM_004004.5(GJB2):c.250G>A (p.Val84Met)	GJB2	13	20189332	Deafness, autosomal dominant 3a;Deafness, autosomal recessive 1A;Nonsyndromic hearing loss and deafness;not provided
NM_004004.5(GJB2):c.428G>A (p.Arg143Gln)	GJB2	13	20189154	Deafness, autosomal dominant 3a;Deafness, autosomal recessive 1A;not provided
NM_004004.5(GJB2):c.551G>A (p.Arg184Gln)	GJB2	13	20189031	Deafness, autosomal dominant 3a;Hearing impairment;not provided
NM_004004.5(GJB2):c.223C>T (p.Arg75Trp)	GJB2	13	20189359	Deafness, autosomal dominant 3a;Nonsyndromic hearing loss and deafness
NM_006783.4(GJB6):c.14C>T (p.Thr5Met)	GJB6	13	20223467	Deafness, autosomal dominant 3b
NM_001145809.1(MYH14):c.3049C>T (p.Leu1017Phe)	MYH14	19	50271424	Deafness, autosomal dominant 4
NM_024729.3(MYH14):c.359C>T (p.Ser120Leu)	MYH14	19	50210724	Deafness, autosomal dominant 4;not provided
NM_001039213.3(CEACAM16):c.505G>A (p.Gly169Arg)	CEACAM16	19	44704140	Deafness, autosomal dominant 4b
NR_029512.1(MIR96):n.13G>A	MIR96	7	129774757	Deafness, autosomal dominant 50
NM_002160.3(TNC):c.5317G>A (p.Val1773Met)	TNC	9	115041016	Deafness, autosomal dominant 56
NM_019887.5(DIABLO):c.377C>T (p.Ser126Leu)	DIABLO	12	122216808	Deafness, autosomal dominant 64
NM_001199107.1(TBC1D24):c.533C>T (p.Ser178Leu)	TBC1D24	16	2496681	Deafness, autosomal dominant 65;Deafness, autosomal recessive 86
NM_006016.5(CD164):c.574C>T (p.Arg192Ter)	CD164	6	109368871	Deafness, autosomal dominant 66
NM_004526.3(MCM2):c.130C>T (p.Arg44Cys)	MCM2	3	127599441	Deafness, autosomal dominant 70

NM_004086.2(COCH):c.263G>A (p.Gly88Glu)	COCH	14	30878834	Deafness, autosomal dominant 9
NM_004086.2(COCH):c.355G>A (p.Ala119Thr)	COCH	14	30878926	Deafness, autosomal dominant 9
NM_004086.2(COCH):c.1625G>A (p.Cys542Tyr)	COCH	14	30889763	Deafness, autosomal dominant 9
NM_004086.2(COCH):c.1159C>T (p.Leu387Phe)	COCH	14	30885994	Deafness, autosomal dominant 9
NM_004086.2(COCH):c.151C>T (p.Pro51Ser)	COCH	14	30877640	Deafness, autosomal dominant 9;Nonsyndromic hearing loss and deafness
NM_002473.5(MYH9):c.287C>T (p.Ser96Leu)	MYH9	22	36348950	Deafness, autosomal dominant nonsyndromic sensorineural 17;Epstein syndrome;MYH9 related disorders
NM_002473.5(MYH9):c.2114G>A (p.Arg705His)	MYH9	22	36305975	Deafness, autosomal dominant nonsyndromic sensorineural 17;MYH9 related disorders
NM_004447.5(EPS8):c.88C>T (p.Gln30Ter)	EPS8	12	15681274	Deafness, autosomal recessive 102
NM_014722.4(RIPOR2):c.102-1G>A	RIPOR2	6	24873800	Deafness, autosomal recessive 104
NM_033312.2(CDC14A):c.1126C>T (p.Arg376Ter)	CDC14A	1	100484440	Deafness, autosomal recessive 105
NM_033312.2(CDC14A):c.1015C>T (p.Arg339Ter)	CDC14A	1	100484329	Deafness, autosomal recessive 105
NM_022124.5(CDH23):c.6133G>A (p.Asp2045Asn)	CDH23	10	71791215	Deafness, autosomal recessive 12
NM_022124.5(CDH23):c.6604G>A (p.Asp2202Asn)	CDH23	10	71793532	Deafness, autosomal recessive 12
NM_022124.5(CDH23):c.719C>T (p.Pro240Leu)	CDH23	10	71570884	Deafness, autosomal recessive 12
NM_001171932.1(CDH23):c.1036C>T (p.Pro346Ser)	CDH23	10	71617295	Deafness, autosomal recessive 12
NM_001171932.1(CDH23):c.1037C>T (p.Pro346Leu)	CDH23	10	71617296	Deafness, autosomal recessive 12
NG_008835.1:g.285670C>T	CDH23	10	71677616	Deafness, autosomal recessive 12
NM_022124.5(CDH23):c.3181G>A (p.Glu1061Lys)	CDH23	10	71709172	Deafness, autosomal recessive 12
NM_022124.5(CDH23):c.5749G>A (p.Glu1917Lys)	CDH23	10	71785667	Deafness, autosomal recessive 12
NM_022124.5(CDH23):c.4021G>A (p.Asp1341Asn)	CDH23	10	71732292	Deafness, autosomal recessive 12;not provided
NM_133261.2(GIPC3):c.903G>A (p.Trp301Ter)	GIPC3	19	3590154	Deafness, autosomal recessive 15
NM_133261.2(GIPC3):c.767G>A (p.Gly256Asp)	GIPC3	19	3589892	Deafness, autosomal recessive 15
NM_133261.2(GIPC3):c.565C>T (p.Arg189Cys)	GIPC3	19	3586967	Deafness, autosomal recessive 15
NM_153700.2(STRC):c.4701+1G>A	STRC	15	43601395	Deafness, autosomal recessive 16;Nonsyndromic hearing loss and deafness
NM_153676.3(USH1C):c.496+1G>A	USH1C	11	17527222	Deafness, autosomal recessive 18;Usher syndrome, type 1C
NM_001277269.1(OTOG):c.6347C>T (p.Pro2116Leu)	OTOG	11	17612638	Deafness, autosomal recessive 18b
NM_001277269.1(OTOG):c.6559C>T (p.Arg2187Ter)	OTOG	11	17613696	Deafness, autosomal recessive 18b
NM_004004.5(GJB2):c.-260C>T	GJB2	13	20193020	Deafness, autosomal recessive 1A
NM_004004.5(GJB2):c.131G>A (p.Trp44Ter)	GJB2	13	20189451	Deafness, autosomal recessive 1A
NM_001097642.2(GJB1):c.131G>A (p.Trp44Ter)	GJB1	X	71223838	Deafness, autosomal recessive 1A
NM_004004.5(GJB2):c.283G>A (p.Val95Met)	GJB2	13	20189299	Deafness, autosomal recessive 1A;Hearing impairment;Nonsyndromic hearing loss and deafness
NM_004004.5(GJB2):c.298C>T (p.His100Tyr)	GJB2	13	20189284	Deafness, autosomal recessive 1A;Hearing impairment;Nonsyndromic hearing loss and deafness
NM_004004.5(GJB2):c.427C>T (p.Arg143Trp)	GJB2	13	20189155	Deafness, autosomal recessive 1A;Hearing impairment;Nonsyndromic hearing loss and deafness;not provided
NM_004004.5(GJB2):c.109G>A (p.Val37Ile)	GJB2	13	20189473	Deafness, autosomal recessive 1A;Hearing impairment;Nonsyndromic hearing loss and deafness;not provided
NM_004004.5(GJB2):c.-23+1G>A	GJB2	13	20192782	Deafness, autosomal recessive 1A;Hearing impairment;Nonsyndromic hearing loss and deafness;not provided
NM_004004.5(GJB2):c.148G>A (p.Asp50Asn)	GJB2	13	20189434	Deafness, autosomal recessive 1A;Hystrix-like ichthyosis with deafness;Keratitis-ichthyosis-deafness syndrome, autosomal dominant;not provided
NM_004004.5(GJB2):c.134G>A (p.Gly45Glu)	GJB2	13	20189448	Deafness, autosomal recessive 1A;Keratitis-ichthyosis-deafness syndrome, autosomal dominant
NM_004004.5(GJB2):c.231G>A (p.Trp77Ter)	GJB2	13	20189351	Deafness, autosomal recessive 1A;Nonsyndromic hearing loss and deafness
NM_004004.5(GJB2):c.370C>T (p.Gln124Ter)	GJB2	13	20189212	Deafness, autosomal recessive 1A;Nonsyndromic hearing loss and deafness
NM_004004.5(GJB2):c.230G>A (p.Trp77Ter)	GJB2	13	20189352	Deafness, autosomal recessive 1A;Nonsyndromic hearing loss and deafness

NM_000260.3(MYO7A):c.1184G>A (p.Arg395His)	MYO7A	11	77160266	Deafness, autosomal recessive 2
NM_000260.3(MYO7A):c.6196C>T (p.Gln2066Ter)	MYO7A	11	77211296	Deafness, autosomal recessive 2
NM_000260.3(MYO7A):c.6211C>T (p.Gln2071Ter)	MYO7A	11	77211311	Deafness, autosomal recessive 2
NM_000260.3(MYO7A):c.6487G>A (p.Gly2163Ser)	MYO7A	11	77213908	Deafness, autosomal recessive 2
NM_000260.3(MYO7A):c.5899C>T (p.Arg1967Ter)	MYO7A	11	77208472	Deafness, autosomal recessive 2;MYO7A-Related Disorders;Retinal dystrophy;Usher syndrome, type 1
NM_000260.3(MYO7A):c.2005C>T (p.Arg669Ter)	MYO7A	11	77174825	Deafness, autosomal recessive 2;Usher syndrome, type 1;not provided
NM_000260.3(MYO7A):c.1797G>A (p.Met599Ile)	MYO7A	11	77166162	Deafness, autosomal recessive 2;Usher syndrome, type 1B
NM_144672.3(OTOA):c.1352G>A (p.Gly451Asp)	OTOA	16	21715016	Deafness, autosomal recessive 22
NM_144672.3(OTOA):c.1879C>T (p.Pro627Ser)	OTOA	16	21722977	Deafness, autosomal recessive 22
NM_033056.3(PCDH15):c.785G>A (p.Gly262Asp)	PCDH15	10	54317362	Deafness, autosomal recessive 23
NM_001142769.1(PCDH15):c.4726C>T (p.Gln1576Ter)	PCDH15	10	53809339	Deafness, autosomal recessive 23
NM_033056.3(PCDH15):c.733C>T (p.Arg245Ter)	PCDH15	10	54317414	Deafness, autosomal recessive 23;Usher syndrome, type 1;Usher syndrome, type 1D;Usher syndrome, type 1F;Usher syndrome, type 1G;not provided
NM_002906.3(RDX):c.1732G>A (p.Asp578Asn)	RDX	11	110231889	Deafness, autosomal recessive 24
NM_002906.3(RDX):c.463C>T (p.Gln155Ter)	RDX	11	110263964	Deafness, autosomal recessive 24
NM_001080476.2(GRXCR1):c.229C>T (p.Gln77Ter)	GRXCR1	4	42893495	Deafness, autosomal recessive 25
NM_001080476.2(GRXCR1):c.412C>T (p.Arg138Cys)	GRXCR1	4	42962919	Deafness, autosomal recessive 25
NM_001039141.2(TRIOBP):c.1039C>T (p.Arg347Ter)	TRIOBP	22	37723595	Deafness, autosomal recessive 28
NM_001039141.2(TRIOBP):c.1741C>T (p.Gln581Ter)	TRIOBP	22	37724297	Deafness, autosomal recessive 28
NM_001039141.2(TRIOBP):c.889C>T (p.Gln297Ter)	TRIOBP	22	37723445	Deafness, autosomal recessive 28
NM_001039141.2(TRIOBP):c.2362C>T (p.Arg788Ter)	TRIOBP	22	37724918	Deafness, autosomal recessive 28
NM_001039141.2(TRIOBP):c.3202C>T (p.Arg1068Ter)	TRIOBP	22	37725758	Deafness, autosomal recessive 28
NM_001039141.2(TRIOBP):c.3349C>T (p.Arg1117Ter)	TRIOBP	22	37725905	Deafness, autosomal recessive 28
NM_001039141.2(TRIOBP):c.1447C>T (p.Gln483Ter)	TRIOBP	22	37724003	Deafness, autosomal recessive 28
NM_001039141.2(TRIOBP):c.3055G>A (p.Gly1019Arg)	TRIOBP	22	37725611	Deafness, autosomal recessive 28
NM_001146077.1(CLDN14):c.301G>A (p.Gly101Arg)	CLDN14	21	36461395	Deafness, autosomal recessive 29
NM_001146077.1(CLDN14):c.242G>A (p.Arg81His)	CLDN14	21	36461454	Deafness, autosomal recessive 29
NM_012130.3(CLDN14):c.167G>A (p.Trp56Ter)	CLDN14	21	36461529	Deafness, autosomal recessive 29
NM_012130.3(CLDN14):c.694G>A (p.Gly232Arg)	CLDN14	21	36461002	Deafness, autosomal recessive 29
NM_016239.3(MYO15A):c.3685C>T (p.Gln1229Ter)	MYO15A	17	18124558	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.6340G>A (p.Val2114Met)	MYO15A	17	18145938	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.8340G>A (p.Thr2780=)	MYO15A	17	18155225	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.8467G>A (p.Asp2823Asn)	MYO15A	17	18156202	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.3944G>A (p.Gly1315Glu)	MYO15A	17	18127077	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.4108C>T (p.Arg1370Cys)	MYO15A	17	18131308	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.5809C>T (p.Arg1937Cys)	MYO15A	17	18142238	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.6589C>T (p.Gln2197Ter)	MYO15A	17	18148108	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.3866+1G>A	MYO15A	17	18126457	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.6178-1G>A	MYO15A	17	18144496	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.4528C>T (p.Gln1510Ter)	MYO15A	17	18135756	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.1223C>T (p.Ala408Val)	MYO15A	17	18120023	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.4240G>A (p.Glu1414Lys)	MYO15A	17	18132486	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.9572G>A (p.Arg3191His)	MYO15A	17	18162639	Deafness, autosomal recessive 3
NM_016239.3(MYO15A):c.8714-1G>A	MYO15A	17	18157155	Deafness, autosomal recessive 3;Nonsyndromic hearing loss and deafness

NM_017433.4(MYO3A):c.2506-1G>A	MYO3A	10	26147429	Deafness, autosomal recessive 30
NM_015404.3(WHRN):c.2332C>T (p.Arg778Ter)	WHRN	9	114403982	Deafness, autosomal recessive 31
NM_015404.3(WHRN):c.1417-1G>A	WHRN	9	114423524	Deafness, autosomal recessive 31
NM_004452.3(ESRRB):c.329C>T (p.Ala110Val)	ESRRB	14	76439682	Deafness, autosomal recessive 35
NM_004452.3(ESRRB):c.545G>A (p.Arg182His)	ESRRB	14	76482046	Deafness, autosomal recessive 35
NM_004452.3(ESRRB):c.787G>A (p.Gly263Ser)	ESRRB	14	76482759	Deafness, autosomal recessive 35
NM_004999.3(MYO6):c.3496C>T (p.Arg1166Ter)	MYO6	6	75914119	Deafness, autosomal recessive 37
NM_000601.5(HGF):c.495G>A (p.Ser165=)	HGF	7	81752250	Deafness, autosomal recessive 39
NM_001199799.1(ILDR1):c.3G>A (p.Met1Ile)	ILDR1	3	122022075	Deafness, autosomal recessive 42
NM_001199799.1(ILDR1):c.583C>T (p.Gln195Ter)	ILDR1	3	122001371	Deafness, autosomal recessive 42
NM_021116.2(ADCY1):c.3112C>T (p.Arg1038Ter)	ADCY1	7	45713747	Deafness, autosomal recessive 44
NM_001042702.3(DFNB59):c.547C>T (p.Arg183Trp)	DFNB59	2	178456149	Deafness, autosomal recessive 59
NM_001042702.3(DFNB59):c.161C>T (p.Thr54Ile)	DFNB59	2	178453570	Deafness, autosomal recessive 59
NM_001042702.3(DFNB59):c.499C>T (p.Arg167Ter)	DFNB59	2	178456101	Deafness, autosomal recessive 59;Nonsyndromic hearing loss and deafness
NM_147196.2(TMIE):c.241C>T (p.Arg81Cys)	TMIE	3	46709155	Deafness, autosomal recessive 6
NM_147196.2(TMIE):c.250C>T (p.Arg84Trp)	TMIE	3	46709164	Deafness, autosomal recessive 6
NM_147196.2(TMIE):c.274C>T (p.Arg92Trp)	TMIE	3	46709188	Deafness, autosomal recessive 6
NM_147196.2(TMIE):c.170G>A (p.Trp57Ter)	TMIE	3	46705866	Deafness, autosomal recessive 6
NM_206883.2(SLC26A5):c.209G>A (p.Trp70Ter)	SLC26A5	7	103420821	Deafness, autosomal recessive 61
NM_001145308.4(LRTOMT):c.242G>A (p.Arg81Gln)	LRTOMT	11	72106094	Deafness, autosomal recessive 63
NM_001145308.4(LRTOMT):c.328G>A (p.Glu110Lys)	LRTOMT	11	72106180	Deafness, autosomal recessive 63
NM_182548.3(LHFPL5):c.494C>T (p.Thr165Met)	LHFPL5	6	35814627	Deafness, autosomal recessive 67
NM_138691.2(TMC1):c.1532C>T (p.Pro511Leu)	TMC1	9	72792318	Deafness, autosomal recessive 7
NM_138691.2(TMC1):c.100C>T (p.Arg34Ter)	TMC1	9	72694578	Deafness, autosomal recessive 7;Nonsyndromic hearing loss and deafness
NM_138691.2(TMC1):c.1165C>T (p.Arg389Ter)	TMC1	9	72789258	Deafness, autosomal recessive 7;Nonsyndromic hearing loss and deafness
NM_001031679.2(MSRB3):c.55C>T (p.Arg19Ter)	MSRB3	12	65308634	Deafness, autosomal recessive 74
NM_144612.6(LOXHD1):c.2008C>T (p.Arg670Ter)	LOXHD1	18	46572125	Deafness, autosomal recessive 77
NM_144612.6(LOXHD1):c.4843G>A (p.Gly1615Arg)	LOXHD1	18	46524499	Deafness, autosomal recessive 77
NM_144612.6(LOXHD1):c.4714C>T (p.Arg1572Ter)	LOXHD1	18	46524734	Deafness, autosomal recessive 77;Nonsyndromic hearing loss and deafness
NM_144612.6(LOXHD1):c.4480C>T (p.Arg1494Ter)	LOXHD1	18	46529227	Deafness, autosomal recessive 77;Nonsyndromic Hearing Loss, Recessive;Nonsyndromic hearing loss and deafness;not provided
NM_001128228.2(TPRN):c.1239G>A (p.Trp413Ter)	TPRN	9	137199473	Deafness, autosomal recessive 79
NM_024022.2(TMPRSS3):c.1211C>T (p.Pro404Leu)	TMPRSS3	21	42375852	Deafness, autosomal recessive 8
NM_024022.2(TMPRSS3):c.325C>T (p.Arg109Trp)	TMPRSS3	21	42388524	Deafness, autosomal recessive 8;Nonsyndromic Hearing Loss, Recessive;Nonsyndromic hearing loss and deafness;not provided
NM_173591.3(OTOGL):c.547C>T (p.Arg183Ter)	OTOGL	12	80229341	Deafness, autosomal recessive 84b
NM_173591.3(OTOGL):c.3076C>T (p.Gln1026Ter)	OTOGL	12	80302673	Deafness, autosomal recessive 84b
NM_001130089.1(KARS):c.1129G>A (p.Asp377Asn)	KARS	16	75631726	Deafness, autosomal recessive 89
NM_194248.2(OTOF):c.2887C>T (p.Arg963Ter)	OTOF	2	26476018	Deafness, autosomal recessive 9
NM_194248.2(OTOF):c.2991+1G>A	OTOF	2	26475913	Deafness, autosomal recessive 9
NM_194248.2(OTOF):c.4275G>A (p.Trp1425Ter)	OTOF	2	26467186	Deafness, autosomal recessive 9
NM_194248.2(OTOF):c.4559G>A (p.Arg1520Gln)	OTOF	2	26466018	Deafness, autosomal recessive 9
NM_194248.2(OTOF):c.5712+1G>A	OTOF	2	26460851	Deafness, autosomal recessive 9
NM_194248.2(OTOF):c.1621G>A (p.Gly541Ser)	OTOF	2	26480968	Deafness, autosomal recessive 9
NM_194248.2(OTOF):c.1273C>T (p.Arg425Ter)	OTOF	2	26483581	Deafness, autosomal recessive 9

NM_194248.2(OTOF):c.1607G>A (p.Trp536Ter)	OTOF	2	26480982	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.1780G>A (p.Glu594Lys)	OTOF	2	26480809	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.1841G>A (p.Gly614Glu)	OTOF	2	26480274	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.3400C>T (p.Arg1134Ter)	OTOF	2	26473999	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.4157C>T (p.Thr1386Ile)	OTOF	2	26467435	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.709C>T (p.Arg237Ter)	OTOF	2	26502301	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.5197G>A (p.Glu1733Lys)	OTOF	2	26462177	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.5567G>A (p.Arg1856Gln)	OTOF	2	26460997	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.5815C>T (p.Arg1939Trp)	OTOF	2	26460204	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.4747C>T (p.Arg1583Cys)	OTOF	2	26465724	Deafness, autosomal recessive 9	
NM_004802.3(OTOF):c.1729C>T (p.Arg577Ter)	OTOF	2	26468468	Deafness, autosomal recessive 9	
NM_194248.2(OTOF):c.4483C>T (p.Arg1495Ter)	OTOF	2	26466731	Deafness, autosomal recessive 9;Hearing impairment	
NM_194248.2(OTOF):c.2122C>T (p.Arg708Ter)	OTOF	2	26479356	Deafness, autosomal recessive 9;Nonsyndromic hearing loss and deafness	
NM_194248.2(OTOF):c.2485C>T (p.Gln829Ter)	OTOF	2	26477210	Deafness, autosomal recessive 9;Nonsyndromic hearing loss and deafness;not provided	
NM_000214.2(JAG1):c.701G>A (p.Cys234Tyr)	JAG1	20	10656452	Deafness, congenital heart defects, and posterior embryotoxon	
NM_001693.3(ATP6V1B2):c.1516C>T (p.Arg506Ter)	ATP6V1B2	8	20220382	Deafness, congenital, with onychodystrophy, autosomal dominant	
NM_001038603.2(MARVELD2):c.1498C>T (p.Arg500Ter)	MARVELD2	5	69433088	Deafness, neurosensory, autosomal recessive 49;Nonsyndromic hearing loss and deafness;not provided	
NM_031475.2(ESPN):c.2230G>A (p.Asp744Asn)	ESPN	1	6452001	Deafness, without vestibular involvement, autosomal dominant	
NM_031475.2(ESPN):c.2321G>A (p.Arg774Gln)	ESPN	1	6452092	Deafness, without vestibular involvement, autosomal dominant	
NM_002764.3(PRPS1):c.193G>A (p.Asp65Asn)	PRPS1	X	107639365	Deafness, X-linked 1	
NM_002764.3(PRPS1):c.259G>A (p.Ala87Thr)	PRPS1	X	107639431	Deafness, X-linked 1	
NM_002764.3(PRPS1):c.916G>A (p.Gly306Arg)	PRPS1	X	107649991	Deafness, X-linked 1	
NM_000307.4(POU3F4):c.935C>T (p.Ala312Val)	POU3F4	X	83509259	Deafness, X-linked 2	
NM_000307.4(POU3F4):c.235C>T (p.Gln79Ter)	POU3F4	X	83508559	Deafness, X-linked 2	
NM_004208.3(AIFM1):c.1265G>A (p.Arg422Gln)	AIFM1	X	130136085	Deafness, X-linked 5	
NM_004208.3(AIFM1):c.1264C>T (p.Arg422Trp)	AIFM1	X	130136086	Deafness, X-linked 5	
NM_004208.3(AIFM1):c.1030C>T (p.Leu344Phe)	AIFM1	X	130137123	Deafness, X-linked 5	
NM_004208.3(AIFM1):c.1352G>A (p.Arg451Gln)	AIFM1	X	130133409	Deafness, X-linked 5	
NM_001847.3(COL4A6):c.1771G>A (p.Gly591Ser)	COL4A6	X	108187279	Deafness, X-linked 6	
NM_000432.3(MYL2):c.64G>A (p.Glu22Lys)	MYL2	12	110919133	Death in early adulthood;Familial hypertrophic cardiomyopathy 10;Primary familial hypertrophic cardiomyopathy;not provided	
NM_194279.3(ISCA2):c.229G>A (p.Gly77Ser)	ISCA2	14	74494329	Death in infancy;Failure to thrive;Global developmental delay;High CSF lactic acid;Multiple mitochondrial dysfunctions syndrome;Multiple mitochondrial dysfunctions syndrome 4;Muscular hypotonia of the trunk;Neurodegeration;Optic atrophy;Spastic tetraplegia;not provided	
NM_001609.3(ACADSB):c.1228G>A (p.Gly410Ser)	ACADSB	10	123053160	Deficiency of 2-methylbutyryl-CoA dehydrogenase	
NM_001609.3(ACADSB):c.763C>T (p.Leu255Phe)	ACADSB	10	123043127	Deficiency of 2-methylbutyryl-CoA dehydrogenase	
NM_001609.3(ACADSB):c.303+1G>A	ACADSB	10	123037848	Deficiency of 2-methylbutyryl-CoA dehydrogenase	
NM_001609.3(ACADSB):c.295C>T (p.Gln99Ter)	ACADSB	10	123037839	Deficiency of 2-methylbutyryl-CoA dehydrogenase	
NM_005327.4(HADH):c.118G>A (p.Ala40Thr)	HADH	4	107990050	Deficiency of 3-hydroxyacyl-CoA dehydrogenase	
NM_000019.3(ACAT1):c.1138G>A (p.Ala380Thr)	ACAT1	11	108146334	Deficiency of acetyl-CoA acetyltransferase	
NM_000019.3(ACAT1):c.547G>A (p.Gly183Arg)	ACAT1	11	108139009	Deficiency of acetyl-CoA acetyltransferase	
NM_000019.3(ACAT1):c.814C>T (p.Gln272Ter)	ACAT1	11	108141688	Deficiency of acetyl-CoA acetyltransferase;not provided	
NM_000528.3(MAN2B1):c.2278C>T (p.Arg760Ter)	MAN2B1	19	12649418	Deficiency of alpha-mannosidase	
NM_000528.3(MAN2B1):c.1915C>T (p.Gln639Ter)	MAN2B1	19	12652376	Deficiency of alpha-mannosidase	

NM_000528.3(MAN2B1):c.2165+1G>A	MAN2B1	19	12650103	Deficiency of alpha-mannosidase
NM_000528.3(MAN2B1):c.418C>T (p.Arg140Ter)	MAN2B1	19	12665370	Deficiency of alpha-mannosidase
NM_001082971.1(DDC):c.304G>A (p.Gly102Ser)	DDC	7	50539926	Deficiency of aromatic-L-amino-acid decarboxylase
NM_001082971.1(DDC):c.749C>T (p.Ser250Phe)	DDC	7	50504025	Deficiency of aromatic-L-amino-acid decarboxylase
NM_001082971.1(DDC):c.272C>T (p.Ala91Val)	DDC	7	50539958	Deficiency of aromatic-L-amino-acid decarboxylase
NM_001082971.1(DDC):c.823G>A (p.Ala275Thr)	DDC	7	50499201	Deficiency of aromatic-L-amino-acid decarboxylase
NM_001082971.1(DDC):c.1040G>A (p.Arg347Gln)	DDC	7	50476625	Deficiency of aromatic-L-amino-acid decarboxylase
NM_199186.2(BPGM):c.268C>T (p.Arg90Cys)	BPGM	7	134661775	Deficiency of bisphosphoglycerate mutase
NM_000055.3(BCHE):c.428G>A (p.Gly143Asp)	BCHE	3	165830606	Deficiency of butyrylcholine esterase
NM_000017.3(ACADS):c.268G>A (p.Gly90Ser)	ACADS	12	120737043	Deficiency of butyryl-CoA dehydrogenase
NM_000017.3(ACADS):c.575C>T (p.Ala192Val)	ACADS	12	120737939	Deficiency of butyryl-CoA dehydrogenase
NM_000017.3(ACADS):c.164C>T (p.Pro55Leu)	ACADS	12	120727143	Deficiency of butyryl-CoA dehydrogenase
NM_000017.3(ACADS):c.323G>A (p.Gly108Asp)	ACADS	12	120737098	Deficiency of butyryl-CoA dehydrogenase
NM_000017.3(ACADS):c.136C>T (p.Arg46Trp)	ACADS	12	120727115	Deficiency of butyryl-CoA dehydrogenase;not provided
NM_000017.3(ACADS):c.319C>T (p.Arg107Cys)	ACADS	12	120737094	Deficiency of butyryl-CoA dehydrogenase;not provided
NM_000017.3(ACADS):c.1147C>T (p.Arg383Cys)	ACADS	12	120739356	Deficiency of butyryl-CoA dehydrogenase;not provided
NM_000017.3(ACADS):c.973C>T (p.Arg325Trp)	ACADS	12	120738859	Deficiency of butyryl-CoA dehydrogenase;not provided
NM_000017.3(ACADS):c.1058C>T (p.Ser353Leu)	ACADS	12	120739168	Deficiency of butyryl-CoA dehydrogenase;not provided
NM_000017.3(ACADS):c.1138C>T (p.Arg380Trp)	ACADS	12	120739347	Deficiency of butyryl-CoA dehydrogenase;not provided
NM_000096.3(CP):c.1874G>A (p.Gly625Glu)	CP	3	149186723	Deficiency of ferroxidase
NM_000096.3(CP):c.2675G>A (p.Gly892Glu)	CP	3	149178618	Deficiency of ferroxidase
NM_000096.3(CP):c.2962G>A (p.Gly988Ser)	CP	3	149177896	Deficiency of ferroxidase
NM_000096.3(CP):c.643C>T (p.Arg215Ter)	CP	3	149209349	Deficiency of ferroxidase
NM_000096.3(CP):c.2701C>T (p.Arg901Ter)	CP	3	149178592	Deficiency of ferroxidase
NM_000096.3(CP):c.146+1G>A	CP	3	149221646	Deficiency of ferroxidase
NM_000096.3(CP):c.1865-1G>A	CP	3	149186733	Deficiency of ferroxidase
NM_000096.3(CP):c.2879-1G>A	CP	3	149177980	Deficiency of ferroxidase
NM_000096.3(CP):c.395-1G>A	CP	3	149210380	Deficiency of ferroxidase
NM_000096.3(CP):c.607+1G>A	CP	3	149210166	Deficiency of ferroxidase
NM_000096.3(CP):c.3019-1G>A	CP	3	149176413	Deficiency of ferroxidase;Hemosiderosis, systemic, due to aceruloplasminemia
NM_000096.3(CP):c.2630G>A (p.Trp877Ter)	CP	3	149179587	Deficiency of ferroxidase;Hypoceruloplasminemia
NM_000154.1(GALK1):c.94G>A (p.Val32Met)	GALK1	17	75765043	Deficiency of galactokinase
NM_000154.1(GALK1):c.1144C>T (p.Gln382Ter)	GALK1	17	75758091	Deficiency of galactokinase
NM_000154.1(GALK1):c.593C>T (p.Ala198Val)	GALK1	17	75763032	Deficiency of galactokinase
NM_001205019.1(GK):c.1255C>T (p.Arg419Ter)	GK	X	30720639	Deficiency of glycerol kinase
NM_000156.5(GAMT):c.506G>A (p.Cys169Tyr)	GAMT	19	1398980	Deficiency of guanidinoacetate methyltransferase
NM_000156.5(GAMT):c.327G>A (p.Lys109=)	GAMT	19	1399793	Deficiency of guanidinoacetate methyltransferase;not provided
NM_000156.5(GAMT):c.522G>A (p.Trp174Ter)	GAMT	19	1398964	Deficiency of guanidinoacetate methyltransferase;not provided
NM_153281.1(HYAL1):c.802G>A (p.Glu268Lys)	HYAL1	3	50302155	Deficiency of hyaluronoglucosaminidase
NM_000191.2(HMGCL):c.835G>A (p.Glu279Lys)	HMGCL	1	23804441	Deficiency of hydroxymethylglutaryl-CoA lyase
NM_000191.2(HMGCL):c.122G>A (p.Arg41Gln)	HMGCL	1	23820532	Deficiency of hydroxymethylglutaryl-CoA lyase;not provided
NM_000547.5(TPO):c.1618C>T (p.Arg540Ter)	TPO	2	1487841	Deficiency of iodide peroxidase
NM_000547.5(TPO):c.1768G>A (p.Gly590Ser)	TPO	2	1487991	Deficiency of iodide peroxidase
NM_000547.5(TPO):c.1943G>A (p.Arg648Gln)	TPO	2	1493976	Deficiency of iodide peroxidase
NM_000547.5(TPO):c.2077C>T (p.Arg693Trp)	TPO	2	1496059	Deficiency of iodide peroxidase

NM_000547.5(TPO):c.1994G>A (p.Arg665Gln)	TPO	2	1494027	Deficiency of iodide peroxidase
NM_014384.2(ACAD8):c.988C>T (p.Arg330Trp)	ACAD8	11	134261786	Deficiency of isobutyryl-CoA dehydrogenase
NM_014384.2(ACAD8):c.1129G>A (p.Gly377Ser)	ACAD8	11	134262556	Deficiency of isobutyryl-CoA dehydrogenase;not provided
NM_014384.2(ACAD8):c.384G>A (p.Met128Ile)	ACAD8	11	134258518	Deficiency of isobutyryl-CoA dehydrogenase;not provided
NM_012213.2(MLYCD):c.8G>A (p.Gly3Asp)	MLYCD	16	83899152	Deficiency of malonyl-CoA decarboxylase
NM_004577.3(PSPH):c.94G>A (p.Asp32Asn)	PSPH	7	56021119	Deficiency of phosphoserine phosphatase
NM_004577.3(PSPH):c.103G>A (p.Ala35Thr)	PSPH	7	56021110	Deficiency of phosphoserine phosphatase
NM_170726.2(ALDH4A1):c.1055C>T (p.Ser352Leu)	ALDH4A1	1	18877498	Deficiency of pyrroline-5-carboxylate reductase
NM_144563.2(RPIA):c.404C>T (p.Ala135Val)	RPIA	2	88729279	Deficiency of ribose-5-phosphate isomerase
NM_000497.3(CYP11B1):c.1343G>A (p.Arg448His)	CYP11B1	8	142875012	Deficiency of steroid 11-beta-monooxygenase
NM_000497.3(CYP11B1):c.953C>T (p.Thr318Met)	CYP11B1	8	142876242	Deficiency of steroid 11-beta-monooxygenase
NM_000497.3(CYP11B1):c.1121G>A (p.Arg374Gln)	CYP11B1	8	142875712	Deficiency of steroid 11-beta-monooxygenase
NM_000497.3(CYP11B1):c.347G>A (p.Trp116Ter)	CYP11B1	8	142879080	Deficiency of steroid 11-beta-monooxygenase
NM_000497.3(CYP11B1):c.956C>T (p.Thr319Met)	CYP11B1	8	142875877	Deficiency of steroid 11-beta-monooxygenase
NM_000102.3(CYP17A1):c.1040G>A (p.Arg347His)	CYP17A1	10	102832610	Deficiency of steroid 17-alpha-monooxygenase;Isolated 17,20-lyase deficiency
NM_000155.3(GALT):c.997C>T (p.Arg333Trp)	GALT	9	34649502	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.130G>A (p.Val44Met)	GALT	9	34647136	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.607G>A (p.Glu203Lys)	GALT	9	34648376	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.25C>T (p.Gln9Ter)	GALT	9	34646729	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.98G>A (p.Arg33His)	GALT	9	34647104	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.160C>T (p.Gln54Ter)	GALT	9	34647166	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.197C>T (p.Pro66Leu)	GALT	9	34647203	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.199C>T (p.Arg67Cys)	GALT	9	34647205	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.238C>T (p.Arg80Ter)	GALT	9	34647244	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.292G>A (p.Asp98Asn)	GALT	9	34647531	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.337G>A (p.Asp113Asn)	GALT	9	34647665	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.368G>A (p.Arg123Gln)	GALT	9	34647696	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.413C>T (p.Thr138Met)	GALT	9	34647867	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.443G>A (p.Arg148Gln)	GALT	9	34647897	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.462G>A (p.Trp154Ter)	GALT	9	34647916	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.490C>T (p.Gln164Ter)	GALT	9	34647944	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.524G>A (p.Gly175Asp)	GALT	9	34648131	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.536G>A (p.Gly179Asp)	GALT	9	34648143	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.542C>T (p.Ser181Phe)	GALT	9	34648149	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.553C>T (p.Pro185Ser)	GALT	9	34648160	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.554C>T (p.Pro185Leu)	GALT	9	34648161	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.564+1G>A	GALT	9	34648172	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.575G>A (p.Ser192Asn)	GALT	9	34648344	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.595G>A (p.Ala199Thr)	GALT	9	34648364	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.601C>T (p.Arg201Cys)	GALT	9	34648370	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.602G>A (p.Arg201His)	GALT	9	34648371	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.610C>T (p.Arg204Ter)	GALT	9	34648379	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.619C>T (p.Gln207Ter)	GALT	9	34648388	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.634C>T (p.Gln212Ter)	GALT	9	34648403	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.658G>A (p.Glu220Lys)	GALT	9	34648427	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase

NM_000155.3(GALT):c.692G>A (p.Arg231His)	GALT	9	34648766	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.747G>A (p.Trp249Ter)	GALT	9	34648821	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.770C>T (p.Pro257Leu)	GALT	9	34648844	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.865C>T (p.Leu289Phe)	GALT	9	34649042	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.871G>A (p.Glu291Lys)	GALT	9	34649048	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.904+5G>A	GALT	9	34649086	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.922G>A (p.Glu308Lys)	GALT	9	34649427	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.947G>A (p.Trp316Ter)	GALT	9	34649452	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.948G>A (p.Trp316Ter)	GALT	9	34649453	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.961C>T (p.His321Tyr)	GALT	9	34649466	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.970C>T (p.Pro324Ser)	GALT	9	34649475	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.974C>T (p.Pro325Leu)	GALT	9	34649479	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.983G>A (p.Arg328His)	GALT	9	34649488	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.986C>T (p.Ser329Phe)	GALT	9	34649491	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.989C>T (p.Ala330Val)	GALT	9	34649494	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.1018G>A (p.Glu340Lys)	GALT	9	34649523	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.1057C>T (p.Gln353Ter)	GALT	9	34649562	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.1059+56C>T	GALT	9	34649620	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.1060-1G>A	GALT	9	34650368	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.1108C>T (p.Gln370Ter)	GALT	9	34650417	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.152G>A (p.Arg51Gln)	GALT	9	34647158	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.777G>A (p.Arg259=)	GALT	9	34648851	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.394C>T (p.His132Tyr)	GALT	9	34647848	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.428C>T (p.Ser143Leu)	GALT	9	34647882	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.247G>A (p.Gly83Arg)	GALT	9	34647253	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.998G>A (p.Arg333Gln)	GALT	9	34649503	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.772C>T (p.Arg258Cys)	GALT	9	34648846	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.775C>T (p.Arg259Trp)	GALT	9	34648849	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.905-1G>A	GALT	9	34649409	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
NM_000155.3(GALT):c.404C>T (p.Ser135Leu)	GALT	9	34647858	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase;not provided
NM_000155.3(GALT):c.556C>T (p.His186Tyr)	GALT	9	34648163	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase;not provided
NM_000155.3(GALT):c.691C>T (p.Arg231Cys)	GALT	9	34648765	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase;not provided
NM_000155.3(GALT):c.442C>T (p.Arg148Trp)	GALT	9	34647896	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase;not provided
NM_000379.3(XDH):c.682C>T (p.Arg228Ter)	XDH	2	31386525	Deficiency of xanthine oxidase
NM_000379.3(XDH):c.445C>T (p.Arg149Cys)	XDH	2	31397718	Deficiency of xanthine oxidase
NM_002250.2(KCNN4):c.844G>A (p.Val282Met)	KCNN4	19	43769805	Dehydrated hereditary stomatocytosis 2
NM_002250.2(KCNN4):c.1055G>A (p.Arg352His)	KCNN4	19	43769027	Dehydrated hereditary stomatocytosis 2;not provided
NM_000399.4(EGR2):c.1234G>A (p.Glu412Lys)	EGR2	10	62813404	Dejerine-sottas neuropathy, autosomal dominant
NM_022455.4(NSD1):c.6050G>A (p.Arg2017Gln)	NSD1	5	177283827	Delayed gross motor development;Delayed speech and language development;Global developmental delay;High anterior hairline;High forehead;Hypertelorism;Hypoplasia of the corpus callosum;Increased body weight;Macrocephaly;Osteopenia;Overgrowth;Pointed chin;Preeclampsia;Scoliosis;Sotos syndrome 1;Tall stature;not provided
NM_023110.2(FGFR1):c.2038C>T (p.Gln680Ter)	FGFR1	8	38414569	Delayed puberty;Kallmann syndrome 2
NM_033115.4(TBCK):c.1708+1G>A	TBCK	4	106194717	Delayed reflexes;Dysmorphism;Global developmental delay;Hypotonia, infantile, with psychomotor retardation and characteristic facies 3;Muscular hypotonia;Seizures;Ventral septal defect

NM_005859.4(PURA):c.556C>T (p.Gln186Ter)	PURA	5	140114737	Delayed speech and language development;Global developmental delay;Intellectual disability;Mental retardation, autosomal dominant 31;Neonatal hypotonia;Seizures
NM_000519.3(HBD):c.*199G>A	HBD	11	5232765	delta Thalassemia
NM_000519.3(HBD):c.113G>A (p.Trp38Ter)	HBD	11	5234193	Delta-0-thalassemia
NM_001145661.1(GATA2):c.761C>T (p.Pro254Leu)	GATA2	3	128485837	Dendritic cell, monocyte, B lymphocyte, and natural killer lymphocyte deficiency
NM_000084.4(CLCN5):c.1942C>T (p.Arg648Ter)	CLCN5	X	50090678	Dent disease 1
NM_000084.4(CLCN5):c.1537G>A (p.Gly513Arg)	CLCN5	X	50090118	Dent disease 1
NM_000084.4(CLCN5):c.100C>T (p.Arg34Ter)	CLCN5	X	50070025	Dent disease 1
NM_000084.4(CLCN5):c.836G>A (p.Trp279Ter)	CLCN5	X	50086359	Dent disease 1
NM_000084.4(CLCN5):c.1399C>T (p.Arg467Ter)	CLCN5	X	50088749	Dent disease 1
NM_000084.4(CLCN5):c.2152C>T (p.Arg718Ter)	CLCN5	X	50092130	Dent disease 1
NM_000084.4(CLCN5):c.731C>T (p.Ser244Leu)	CLCN5	X	50085987	Dent disease 1;Hypophosphatemic rickets, X-linked recessive;not provided
NM_000084.4(CLCN5):c.1039C>T (p.Arg347Ter)	CLCN5	X	50086562	Dent disease 1;not provided
NM_000084.4(CLCN5):c.1909C>T (p.Arg637Ter)	CLCN5	X	50090490	Dent disease 1;not provided
NM_001127899.3(CLCN5):c.1047G>A (p.Trp349Ter)	CLCN5	X	50086360	Dent disease 1;Proteinuria, low molecular weight, with hypercalciuria and nephrocalcinosis
NM_000276.3(OCRL):c.952C>T (p.Arg318Cys)	OCRL	X	129562396	Dent disease 2;Lowe syndrome
NM_000276.3(OCRL):c.2299C>T (p.Gln767Ter)	OCRL	X	129588221	Dent disease 2;Lowe syndrome
NM_000276.3(OCRL):c.2582-1G>A	OCRL	X	129590145	Dent disease 2;Lowe syndrome;not provided
NM_014208.3(DSPP):c.133C>T (p.Gln45Ter)	DSPP	4	87612186	Dentinogenesis imperfecta - Shield's type II
NM_014208.3(DSPP):c.44C>T (p.Ala15Val)	DSPP	4	87610952	Dentinogenesis imperfecta - Shield's type II
NM_000169.2(GLA):c.561G>A (p.Met187Ile)	GLA	X	101400744	Deoxygalactonojirimycin response;Fabry disease
NM_000169.2(GLA):c.104G>A (p.Gly35Glu)	GLA	X	101407800	Deoxygalactonojirimycin response;Fabry disease
NM_000302.3(PLOD1):c.1562G>A (p.Trp521Ter)	PLOD1	1	11965571	Depressed nasal bridge;Generalized hypotonia;High palate;Joint hypermobility;Omphalocele;Thoracolumbar scoliosis
NM_014319.4(LEMD3):c.2564G>A (p.Trp855Ter)	LEMD3	12	65245931	Dermatofibrosis lenticularis disseminata
NM_014319.4(LEMD3):c.1963C>T (p.Arg655Ter)	LEMD3	12	65239970	Dermatofibrosis lenticularis disseminata, isolated;Osteopoikilosis with melorheostosis
NM_100264.2(WAC):c.1586G>A (p.Trp529Ter)	WAC	10	28616337	Desanto-shinawi syndrome
NM_100264.2(WAC):c.1717C>T (p.Gln573Ter)	WAC	10	28617762	Desanto-shinawi syndrome
NM_001159772.1(CANT1):c.898C>T (p.Arg300Cys)	CANT1	17	78993858	Desbuquois dysplasia 1
NM_001159772.1(CANT1):c.899G>A (p.Arg300His)	CANT1	17	78993857	Desbuquois dysplasia 1
NM_001159772.1(CANT1):c.374G>A (p.Trp125Ter)	CANT1	17	78997249	Desbuquois dysplasia 1
NM_001159772.1(CANT1):c.896C>T (p.Pro299Leu)	CANT1	17	78993860	Desbuquois dysplasia 1
NM_001159772.1(CANT1):c.676G>A (p.Val226Met)	CANT1	17	78995177	Desbuquois dysplasia 1
NM_001159772.1(CANT1):c.-286+1G>A	CANT1	17	79009663	Desbuquois dysplasia 1
NM_022166.3(XYLT1):c.1441C>T (p.Arg481Trp)	XYLT1	16	17141299	Desbuquois dysplasia 2
NM_022166.3(XYLT1):c.1792C>T (p.Arg598Cys)	XYLT1	16	17134708	Desbuquois dysplasia 2
NM_022166.3(XYLT1):c.439C>T (p.Arg147Ter)	XYLT1	16	17259462	Desbuquois dysplasia 2
NM_022166.3(XYLT1):c.1588-3C>T	XYLT1	16	17138534	Desbuquois dysplasia 2
NM_014762.3(DHCR24):c.571G>A (p.Glu191Lys)	DHCR24	1	54875134	Desmosterolosis
NM_014762.3(DHCR24):c.307C>T (p.Arg103Cys)	DHCR24	1	54883698	Desmosterolosis
NM_014762.3(DHCR24):c.281G>A (p.Arg94His)	DHCR24	1	54883724	Desmosterolosis
NM_014762.3(DHCR24):c.1438G>A (p.Glu480Lys)	DHCR24	1	54852346	Desmosterolosis
NM_004700.3(KCNQ4):c.961G>A (p.Gly321Ser)	KCNQ4	1	40820180	DFNA 2 Nonsyndromic Hearing Loss
NM_004700.3(KCNQ4):c.886G>A (p.Gly296Ser)	KCNQ4	1	40819926	DFNA 2 Nonsyndromic Hearing Loss
NM_004700.3(KCNQ4):c.778G>A (p.Glu260Lys)	KCNQ4	1	40819416	DFNA 2 Nonsyndromic Hearing Loss

NM_004700.3(KCNQ4):c.871C>T (p.Pro291Ser)	KCNQ4	1	40819911	DFNA 2 Nonsyndromic Hearing Loss
NM_004700.3(KCNQ4):c.872C>T (p.Pro291Leu)	KCNQ4	1	40819912	DFNA 2 Nonsyndromic Hearing Loss
NM_004700.3(KCNQ4):c.2039C>T (p.Ser680Phe)	KCNQ4	1	40838474	DFNA 2 Nonsyndromic Hearing Loss
NM_004700.3(KCNQ4):c.853G>A (p.Gly285Ser)	KCNQ4	1	40819893	DFNA 2 Nonsyndromic Hearing Loss;Nonsyndromic hearing loss and deafness
NM_006005.3(WFS1):c.2590G>A (p.Glu864Lys)	WFS1	4	6302385	DFNA6/14/38 Nonsyndromic Low-Frequency Sensorineural Hearing Loss;WFS1-Related Disorders;Wolfram-like syndrome, autosomal dominant
NM_000486.5(AQP2):c.772G>A (p.Glu258Lys)	AQP2	12	49955564	Diabetes insipidus, nephrogenic, autosomal dominant
NM_000486.5(AQP2):c.190G>A (p.Gly64Arg)	AQP2	12	49951020	Diabetes insipidus, nephrogenic, autosomal recessive
NM_000486.5(AQP2):c.377C>T (p.Thr126Met)	AQP2	12	49954171	Diabetes insipidus, nephrogenic, autosomal recessive
NM_000486.5(AQP2):c.523G>A (p.Gly175Arg)	AQP2	12	49954317	Diabetes insipidus, nephrogenic, autosomal recessive
NM_000486.5(AQP2):c.374C>T (p.Thr125Met)	AQP2	12	49954168	Diabetes insipidus, nephrogenic, autosomal recessive
NM_000486.5(AQP2):c.785C>T (p.Pro262Leu)	AQP2	12	49955577	Diabetes insipidus, nephrogenic, autosomal recessive
NM_000486.5(AQP2):c.568G>A (p.Ala190Thr)	AQP2	12	49954672	Diabetes insipidus, nephrogenic, autosomal recessive
NM_000486.5(AQP2):c.559C>T (p.Arg187Cys)	AQP2	12	49954663	Diabetes insipidus, nephrogenic, autosomal recessive;Nephrogenic diabetes insipidus
NM_000486.5(AQP2):c.439G>A (p.Ala147Thr)	AQP2	12	49954233	Diabetes insipidus, nephrogenic, autosomal recessive;Nephrogenic diabetes insipidus
NM_000490.4(AVP):c.20C>T (p.Pro7Leu)	AVP	20	3084655	Diabetes insipidus, neurohypophyseal, autosomal recessive
NM_000525.3(KCNJ11):c.685G>A (p.Glu229Lys)	KCNJ11	11	17387407	Diabetes mellitus
NM_006005.3(WFS1):c.2171C>T (p.Pro724Leu)	WFS1	4	6301966	Diabetes mellitus AND insipidus with optic atrophy AND deafness
NM_006005.3(WFS1):c.1944G>A (p.Trp648Ter)	WFS1	4	6301739	Diabetes mellitus AND insipidus with optic atrophy AND deafness
NM_006005.3(WFS1):c.1511C>T (p.Pro504Leu)	WFS1	4	6301306	Diabetes mellitus AND insipidus with optic atrophy AND deafness
NM_006005.3(WFS1):c.676C>T (p.Gln226Ter)	WFS1	4	6291961	Diabetes mellitus AND insipidus with optic atrophy AND deafness
NM_006005.3(WFS1):c.2455C>T (p.Gln819Ter)	WFS1	4	6302250	Diabetes mellitus AND insipidus with optic atrophy AND deafness
NM_006005.3(WFS1):c.124C>T (p.Arg42Ter)	WFS1	4	6277579	Diabetes mellitus AND insipidus with optic atrophy AND deafness
NM_006005.3(WFS1):c.2146G>A (p.Ala716Thr)	WFS1	4	6301941	Diabetes mellitus AND insipidus with optic atrophy AND deafness;WFS1-Related Disorders
NM_006005.3(WFS1):c.2051C>T (p.Ala684Val)	WFS1	4	6301846	Diabetes mellitus AND insipidus with optic atrophy AND deafness;Wolfram-like syndrome, autosomal dominant;not provided
NM_023083.3(CAPN10):c.471-176G>A	CAPN10	2	240591757	Diabetes mellitus type 2
NC_000002.12:g.240603286C>T	CAPN10	2	240603286	Diabetes mellitus type 2
NM_005456.3(MAPK8IP1):c.176G>A (p.Ser59Asn)	MAPK8IP1	11	45898159	Diabetes mellitus type 2
NM_006193.2(PAX4):c.361C>T (p.Arg121Trp)	PAX4	7	127614533	Diabetes mellitus type 2
NM_001626.5(AKT2):c.821G>A (p.Arg274His)	AKT2	19	40237979	Diabetes mellitus type 2
NM_000208.3(INSR):c.3572G>A (p.Arg1191Gln)	INSR	19	7120707	Diabetes mellitus type 2
NM_000160.4(GCGR):c.118G>A (p.Gly40Ser)	GCGR	17	81809839	Diabetes mellitus type 2
NM_173851.2(SLC30A8):c.412C>T (p.Arg138Ter)	SLC30A8	8	117153084	Diabetes mellitus type 2
NM_000162.3(GCK):c.544G>A (p.Val182Met)	GCK	7	44150004	Diabetes mellitus type 2;Diabetes mellitus, gestational;Maturity-onset diabetes of the young, type 2;not provided
NM_001287174.1(ABCC8):c.4138C>T (p.Arg1380Cys)	ABCC8	11	17395915	Diabetes mellitus type 2;Transient neonatal diabetes mellitus 2
NM_000162.3(GCK):c.706G>A (p.Glu236Lys)	GCK	7	44147807	Diabetes mellitus, gestational
NM_000207.2(INS):c.163C>T (p.Arg55Cys)	INS	11	2160809	Diabetes mellitus, insulin-dependent, 2
NM_000207.2(INS):c.94G>A (p.Gly32Ser)	INS;INS-IGF2	11	2160878	Diabetes mellitus, insulin-dependent, 2;Neonatal diabetes mellitus;Permanent neonatal diabetes mellitus
NM_000545.6(HNF1A):c.815G>A (p.Arg272His)	HNF1A	12	120994265	Diabetes mellitus, insulin-dependent, 20;Monogenic diabetes;not provided
NM_001002255.1(SUMO4):c.163G>A (p.Val55Met)	SUMO4	6	149400554	Diabetes mellitus, insulin-dependent, 5
NM_001042413.1(GLIS3):c.1681C>T (p.His561Tyr)	GLIS3	9	4117797	Diabetes mellitus, neonatal, with congenital hypothyroidism
NM_014832.4(TBC1D4):c.2050C>T (p.Arg684Ter)	TBC1D4	13	75324385	Diabetes mellitus, noninsulin-dependent, 5

NM_000162.3(GCK):c.556C>T (p.Arg186Ter)	GCK	7	44149992	Diabetes mellitus, noninsulin-dependent, late-onset
m.3256C>T	MT-TL1	MT	3256	Diabetes mellitus, noninsulin-dependent, maternally transmitted;Myoclonus with epilepsy with ragged red fibers
NM_178161.2(PTF1A):c.886C>T (p.Arg296Ter)	PTF1A	10	23193805	Diabetes mellitus, permanent neonatal, with cerebellar agenesis
NM_000525.3(KCNJ11):c.175G>A (p.Val59Met)	KCNJ11	11	17387917	Diabetes mellitus, permanent neonatal, with neurologic features;Neonatal insulin-dependent diabetes mellitus;Permanent neonatal diabetes mellitus
NM_000525.3(KCNJ11):c.602G>A (p.Arg201His)	KCNJ11	11	17387490	Diabetes mellitus, permanent neonatal, with neurologic features;Neonatal insulin-dependent diabetes mellitus;Permanent neonatal diabetes mellitus;Transient neonatal diabetes mellitus 3
NM_000525.3(KCNJ11):c.158G>A (p.Gly53Asp)	KCNJ11	11	17387934	Diabetes mellitus, permanent neonatal, with neurologic features;Permanent neonatal diabetes mellitus
NM_000525.3(KCNJ11):c.601C>T (p.Arg201Cys)	KCNJ11	11	17387491	Diabetes mellitus;Diabetes mellitus, permanent neonatal, with neurologic features;Permanent neonatal diabetes mellitus
NM_000525.3(KCNJ11):c.679G>A (p.Glu227Lys)	KCNJ11	11	17387413	Diabetes mellitus;Maturity-onset diabetes of the young, type 13
NM_000525.3(KCNJ11):c.964G>A (p.Glu322Lys)	KCNJ11	11	17387128	Diabetes mellitus;Permanent neonatal diabetes mellitus
NM_001022.3(RPS19):c.184C>T (p.Arg62Trp)	RPS19	19	41869042	Diamond-Blackfan anemia 1
NM_001022.3(RPS19):c.98G>A (p.Trp33Ter)	RPS19	19	41861138	Diamond-Blackfan anemia 1
NM_001022.3(RPS19):c.380G>A (p.Gly127Glu)	RPS19	19	41869722	Diamond-Blackfan anemia 1
NM_001022.3(RPS19):c.280C>T (p.Arg94Ter)	RPS19	19	41869138	Diamond-Blackfan anemia 1;not provided
NM_001029.3(RPS26):c.97G>A (p.Asp33Asn)	RPS26	12	56042518	Diamond-Blackfan anemia 10
NM_001029.3(RPS26):c.3+1G>A	RPS26	12	56042170	Diamond-Blackfan anemia 10
NM_001029.3(RPS26):c.55C>T (p.Gln19Ter)	RPS26	12	56042476	Diamond-Blackfan anemia 10
NM_001029.3(RPS26):c.259C>T (p.Arg87Ter)	RPS26	12	56043440	Diamond-Blackfan anemia 10;Diamond-Blackfan anemia 15 with mandibulofacial dysostosis
NM_033022.3(RPS24):c.316C>T (p.Gln106Ter)	RPS24	10	78037230	Diamond-Blackfan anemia 3
NM_033022.3(RPS24):c.46C>T (p.Arg16Ter)	RPS24	10	78035394	Diamond-Blackfan anemia 3
NM_000996.2(RPL35A):c.97G>A (p.Val33Ile)	RPL35A	3	197951244	Diamond-Blackfan anemia 5
NM_000996.2(RPL35A):c.304C>T (p.Arg102Ter)	RPL35A	3	197954142	Diamond-Blackfan anemia 5
NM_000975.3(RPL11):c.223C>T (p.Arg75Ter)	RPL11	1	23693872	Diamond-Blackfan anemia 7
NM_001011.3(RPS7):c.147+1G>A	RPS7	2	3575889	Diamond-Blackfan anemia 8
NM_001014.4(RPS10):c.3G>A (p.Met1Ile)	RPS10	6	34425219	Diamond-Blackfan anemia 9
NM_001014.4(RPS10):c.337C>T (p.Arg113Ter)	RPS10	6	34421793	Diamond-Blackfan anemia 9
NM_133468.4(BMPER):c.925C>T (p.Gln309Ter)	BMPER	7	34055301	Diaphanospondylodysostosis
NM_133468.4(BMPER):c.1109C>T (p.Pro370Leu)	BMPER	7	34078887	Diaphanospondylodysostosis
NM_012082.3(ZFPM2):c.334C>T (p.Arg112Ter)	ZFPM2	8	105561395	Diaphragmatic hernia 3
NM_000660.6(TGFB1):c.652C>T (p.Arg218Cys)	TGFB1	19	41342230	Diaphyseal dysplasia
NM_000660.6(TGFB1):c.653G>A (p.Arg218His)	TGFB1	19	41342229	Diaphyseal dysplasia
NM_021102.3(SPINT2):c.593-1G>A	SPINT2	19	38291839	Diarrhea 3, secretory sodium, congenital, syndromic
NM_021102.3(SPINT2):c.502G>A (p.Gly168Ser)	SPINT2	19	38290229	Diarrhea 3, secretory sodium, congenital, syndromic
NM_002354.2(EPCAM):c.491+1G>A	EPCAM	2	47375300	Diarrhea 5, with tufting enteropathy, congenital
NM_002354.2(EPCAM):c.197G>A (p.Cys66Tyr)	EPCAM	2	47373820	Diarrhea 5, with tufting enteropathy, congenital
NM_002354.2(EPCAM):c.412C>T (p.Arg138Ter)	EPCAM	2	47374035	Diarrhea 5, with tufting enteropathy, congenital
NM_004174.3(SLC9A3):c.932C>T (p.Ala311Val)	SLC9A3	5	484520	Diarrhea 8, secretory sodium, congenital
NM_004174.3(SLC9A3):c.1145G>A (p.Arg382Gln)	SLC9A3	5	483270	Diarrhea 8, secretory sodium, congenital
NM_004174.3(SLC9A3):c.805G>A (p.Ala269Thr)	SLC9A3	5	484647	Diarrhea 8, secretory sodium, congenital
NM_000112.3(SLC26A2):c.398C>T (p.Ala133Val)	SLC26A2	5	149978050	Diastrophic dysplasia
NM_004170.5(SLC1A1):c.1333C>T (p.Arg445Trp)	SLC1A1	9	4585316	Dicarboxylic aminoaciduria

NM_177438.2(DICER1):c.559C>T (p.Arg187Ter)	DICER1	14	95130072	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.2026C>T (p.Arg676Ter)	DICER1	14	95113106	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.5123G>A (p.G1708E)	DICER1	14	95094129	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.5104C>T (p.Q1702*)	DICER1	14	95094148	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.5053C>T (p.Q1685*)	DICER1	14	95095867	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.4517G>A (p.W1506*)	DICER1	14	95096403	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.4190G>A (p.W1397*)	DICER1	14	95099796	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.3658C>T (p.Q1220*)	DICER1	14	95103738	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.1966C>T (p.R656*)	DICER1	14	95113166	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.1870C>T (p.R624*)	DICER1	14	95115704	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.1525C>T (p.R509*)	DICER1	14	95116680	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.832C>T (p.L278F)	DICER1	14	95126651	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.745C>T (p.G249*)	DICER1	14	95126738	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.3007C>T (p.Arg1003Ter)	DICER1	14	95105764	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.2233C>T (p.Arg745Ter)	DICER1	14	95111340	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.5494C>T (p.Gln1832Ter)	DICER1	14	95091236	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
NM_177438.2(DICER1):c.2062C>T (p.R688*)	DICER1	14	95112226	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome;not provided
NM_177438.2(DICER1):c.2830C>T (p.Arg944Ter)	DICER1	14	95106198	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome;Pleuropulmonary blastoma
NM_177438.2(DICER1):c.1630C>T (p.Arg544Ter)	DICER1	14	95116575	DICER1-related pleuropulmonary blastoma cancer predisposition syndrome;Pleuropulmonary blastoma
NM_000026.3(ADSL):c.421C>T (p.Arg141Trp)	ADSL	22	40354266	Difficulty standing;Generalized myoclonic seizures;inability to walk;Progressive neurologic deterioration;Severe global developmental delay;not provided
NM_024426.4(WT1):c.1390G>A (p.Asp464Asn)	WT1	11	32392014	Diffuse mesangial sclerosis;Drash syndrome
NM_024426.4(WT1):c.1333C>T (p.His445Tyr)	WT1	11	32392672	Diffuse mesangial sclerosis;Drash syndrome
NM_024426.4(WT1):c.1384C>T (p.Arg462Trp)	WT1	11	32392020	Diffuse mesangial sclerosis;Drash syndrome;Frasier syndrome;Meacham syndrome;Wilms tumor 1;not provided
NM_024426.4(WT1):c.1432+4C>T	WT1	11	32391968	Diffuse mesangial sclerosis;Frasier syndrome;Hereditary nephrotic syndrome
NM_001651.3(AQP5):c.562C>T (p.Arg188Cys)	AQP5	12	49964125	Diffuse palmoplantar keratoderma, Bothnian type
NM_080647.1(TBX1):c.928G>A (p.Gly310Ser)	TBX1	22	19765921	DiGeorge sequence
NM_001199107.1(TBC1D24):c.118C>T (p.Arg40Cys)	TBC1D24	16	2496266	Digitorenocerebral syndrome
NM_001199107.1(TBC1D24):c.1206+5G>A	TBC1D24	16	2499425	Digitorenocerebral syndrome
NM_001199107.1(TBC1D24):c.724C>T (p.Arg242Cys)	TBC1D24	16	2496872	Digitorenocerebral syndrome;not provided
NM_001199107.1(TBC1D24):c.328G>A (p.Gly110Ser)	TBC1D24	16	2496476	Digitorenocerebral syndrome;not provided
NM_000320.2(QDPR):c.68G>A (p.Gly23Asp)	QDPR	4	17511987	Dihydropteridine reductase deficiency
NM_000320.2(QDPR):c.270G>A (p.Trp90Ter)	QDPR	4	17504404	Dihydropteridine reductase deficiency
NM_001385.2(DPYS):c.1303G>A (p.Gly435Arg)	DPYS	8	104392924	Dihydropyrimidinase deficiency
NM_001385.2(DPYS):c.424-1G>A	DPYS	8	104447504	Dihydropyrimidinase deficiency
NM_000110.3(DPYD):c.2657G>A (p.Arg886His)	DPYD	1	97098598	Dihydropyrimidine dehydrogenase deficiency
NM_005572.3(LMNA):c.1228C>T (p.Gln410Ter)	LMNA	1	156136284	Dilated cardiomyopathy 1A
NM_170707.3(LMNA):c.1130G>A (p.Arg377His)	LMNA	1	156136094	Dilated cardiomyopathy 1A;Limb-girdle muscular dystrophy, type 1B;not provided
NM_170707.3(LMNA):c.29C>T (p.Thr10Ile)	LMNA	1	156114947	Dilated cardiomyopathy 1A;not provided
NM_170707.3(LMNA):c.568C>T (p.Arg190Trp)	LMNA	1	156134457	Dilated cardiomyopathy 1A;not provided
NM_170707.3(LMNA):c.481G>A (p.Glu161Lys)	LMNA	1	156130741	Dilated cardiomyopathy 1A;Primary dilated cardiomyopathy;not provided
NM_170707.3(LMNA):c.607G>A (p.Glu203Lys)	LMNA	1	156134496	Dilated cardiomyopathy 1A;Primary dilated cardiomyopathy;not provided

NM_007078.2(LDB3):c.617C>T (p.Thr206Ile)	LDB3	10	86681731	Dilated cardiomyopathy 1C
NM_001001430.2(TNNT2):c.430C>T (p.Arg144Trp)	TNNT2	1	201364327	Dilated cardiomyopathy 1DD
NM_001134363.2(RBM20):c.1901G>A (p.Arg634Gln)	RBM20	10	110812298	Dilated cardiomyopathy 1DD;not provided
NM_001134363.2(RBM20):c.1913C>T (p.Pro638Leu)	RBM20	10	110812310	Dilated cardiomyopathy 1DD;Primary dilated cardiomyopathy;not provided
NM_001134363.2(RBM20):c.1907G>A (p.Arg636His)	RBM20	10	110812304	Dilated cardiomyopathy 1DD;Primary dilated cardiomyopathy;not provided
NM_002471.3(MYH6):c.2489C>T (p.Pro830Leu)	MYH6	14	23394264	Dilated cardiomyopathy 1EE
NM_002471.3(MYH6):c.4369G>A (p.Glu1457Lys)	MYH6	14	23387914	Dilated cardiomyopathy 1EE
NM_000363.4(TNNI3):c.485G>A (p.Arg162Gln)	TNNI3	19	55154094	Dilated cardiomyopathy 1FF;Dilated cardiomyopathy 2A;Familial hypertrophic cardiomyopathy 7;Familial restrictive cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_001256850.1(TTN):c.2228C>T (p.Ala743Val)	TTN	2	178785990	Dilated cardiomyopathy 1G
NM_001256850.1(TTN):c.160G>A (p.Val54Met)	TTN	2	178802273	Dilated cardiomyopathy 1G
NM_001256850.1(TTN):c.12157C>T (p.Gln4053Ter)	TTN	2	178740125	Dilated cardiomyopathy 1G
NM_003319.4(TTN):c.13250G>A (p.Ser4417Asn)	TTN	2	178738114	Dilated cardiomyopathy 1G
NM_001256850.1(TTN):c.76114C>T (p.Arg25372Ter)	TTN	2	178565095	Dilated cardiomyopathy 1G;Distal myopathy Markesbery-Griggs type;Familial hypertrophic cardiomyopathy 9;Hereditary myopathy with early respiratory failure;Limb-girdle muscular dystrophy, type 2J;Myopathy, early-onset, with fatal cardiomyopathy;not provided
NM_001256850.1(TTN):c.77113C>T (p.Gln25705Ter)	TTN	2	178564096	Dilated cardiomyopathy 1G;Distal myopathy Markesbery-Griggs type;Familial hypertrophic cardiomyopathy 9;Hereditary myopathy with early respiratory failure;Limb-girdle muscular dystrophy, type 2J;Myopathy, early-onset, with fatal cardiomyopathy;not provided
NM_001256850.1(TTN):c.56953C>T (p.Arg18985Ter)	TTN	2	178589849	Dilated cardiomyopathy 1G;Distal myopathy Markesbery-Griggs type;Familial hypertrophic cardiomyopathy 9;Hereditary myopathy with early respiratory failure;Limb-girdle muscular dystrophy, type 2J;Myopathy, early-onset, with fatal cardiomyopathy;Primary dilated cardiomyopathy;not provided
NM_001267550.2(TTN):c.67495C>T (p.Arg22499Ter)	TTN	2	178579702	Dilated cardiomyopathy 1G;Distal myopathy Markesbery-Griggs type;Familial hypertrophic cardiomyopathy 9;Hereditary myopathy with early respiratory failure;Limb-girdle muscular dystrophy, type 2J;Myopathy, early-onset, with fatal cardiomyopathy;Primary dilated cardiomyopathy;not provided
NM_001267550.2(TTN):c.83515C>T (p.Arg27839Ter)	TTN	2	178562617	Dilated cardiomyopathy 1G;Primary dilated cardiomyopathy
NM_001256850.1(TTN):c.69415C>T (p.Arg23139Ter)	TTN	2	178571794	Dilated cardiomyopathy 1G;Primary dilated cardiomyopathy;not provided
NM_004168.3(SDHA):c.1664G>A (p.Gly555Glu)	SDHA	5	251338	Dilated cardiomyopathy 1GG;Mitochondrial complex II deficiency
NM_004281.3(BAG3):c.1430G>A (p.Arg477His)	BAG3	10	119676984	Dilated cardiomyopathy 1HH
NM_004281.3(BAG3):c.268C>T (p.Arg90Ter)	BAG3	10	119669938	Dilated cardiomyopathy 1HH;not provided
NM_004281.3(BAG3):c.925C>T (p.Arg309Ter)	BAG3	10	119676479	Dilated cardiomyopathy 1HH;Primary dilated cardiomyopathy;not provided
NM_001927.3(DES):c.735+1G>A	DES	2	219420347	Dilated cardiomyopathy 1I;Myofibrillar myopathy 1
NM_001927.3(DES):c.1360C>T (p.Arg454Trp)	DES	2	219425734	Dilated cardiomyopathy 1I;Myofibrillar myopathy 1;Primary familial hypertrophic cardiomyopathy;not provided
NM_001885.2(CRYAB):c.470G>A (p.Arg157His)	CRYAB	11	111908822	Dilated cardiomyopathy 1II
NM_002290.4(LAMA4):c.3217C>T (p.Arg1073Ter)	LAMA4	6	112139164	Dilated cardiomyopathy 1JJ
NM_002290.4(LAMA4):c.2828C>T (p.Pro943Leu)	LAMA4	6	112140887	Dilated cardiomyopathy 1JJ
NM_032578.3(MYPN):c.3263G>A (p.Arg1088His)	MYPN	10	68197456	Dilated cardiomyopathy 1KK;not provided
NM_022114.3(PRDM16):c.872C>T (p.Pro291Leu)	PRDM16	1	3402986	Dilated cardiomyopathy 1LL
NM_003673.3(TCAP):c.410C>T (p.Thr137Ile)	TCAP	17	39666015	Dilated cardiomyopathy 1N
NM_003673.3(TCAP):c.157C>T (p.Gln53Ter)	TCAP	17	39665762	Dilated cardiomyopathy 1N;Limb-girdle muscular dystrophy, type 2G;Primary dilated cardiomyopathy
NM_020297.3(ABCC9):c.4537G>A (p.Ala1513Thr)	ABCC9	12	21801157	Dilated cardiomyopathy 1O

NM_002667.4(PLN):c.25C>T (p.Arg9Cys)	PLN	6	118558946	Dilated cardiomyopathy 1P;Primary dilated cardiomyopathy;not provided
NM_005159.4(ACTC1):c.941G>A (p.Arg314His)	ACTC1	15	34791163	Dilated cardiomyopathy 1R
NM_000257.3(MYH7):c.667G>A (p.Ala223Thr)	MYH7	14	23431650	Dilated cardiomyopathy 1S
NM_000257.3(MYH7):c.1925C>T (p.Ser642Leu)	MYH7	14	23427271	Dilated cardiomyopathy 1S
NM_000257.3(MYH7):c.2710C>T (p.Arg904Cys)	MYH7	14	23424119	Dilated cardiomyopathy 1S;Primary dilated cardiomyopathy
NM_014000.2(VCL):c.2923C>T (p.Arg975Trp)	VCL	10	74112086	Dilated cardiomyopathy 1W;Familial hypertrophic cardiomyopathy 15
NM_001018005.1(TPM1):c.118G>A (p.Glu40Lys)	TPM1	15	63044030	Dilated cardiomyopathy 1Y
NM_001018005.1(TPM1):c.160G>A (p.Glu54Lys)	TPM1	15	63044072	Dilated cardiomyopathy 1Y;not provided
NM_001018005.1(TPM1):c.688G>A (p.Asp230Asn)	TPM1	15	63062263	Dilated cardiomyopathy 1Y;Primary dilated cardiomyopathy;not provided
NM_003280.2(TNNC1):c.476G>A (p.Gly159Asp)	TNNC1	3	52451285	Dilated cardiomyopathy 1Z
NM_004006.2(DMD):c.10192C>T (p.Gln3398Ter)	DMD	X	31178700	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.2137C>T (p.Gln713Ter)	DMD	X	32545190	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.2436G>A (p.Trp812Ter)	DMD	X	32491463	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.2622+1G>A	DMD	X	32491276	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.2758C>T (p.Gln920Ter)	DMD	X	32484964	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.2803+1G>A	DMD	X	32484918	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.2866C>T (p.Gln956Ter)	DMD	X	32472247	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.2977C>T (p.Gln993Ter)	DMD	X	32468683	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.3087G>A (p.Trp1029Ter)	DMD	X	32468573	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.3433-1G>A	DMD	X	32454833	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.4405C>T (p.Gln1469Ter)	DMD	X	32389614	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.4518+5G>A	DMD	X	32389496	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.489G>A (p.Trp163Ter)	DMD	X	32816509	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.4996C>T (p.Arg1666Ter)	DMD	X	32365049	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.5134C>T (p.Gln1712Ter)	DMD	X	32364602	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.5353C>T (p.Gln1785Ter)	DMD	X	32348501	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.5554C>T (p.Gln1852Ter)	DMD	X	32345975	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.6283C>T (p.Arg2095Ter)	DMD	X	32287536	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.7229G>A (p.Trp2410Ter)	DMD	X	31820055	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.7818G>A (p.Trp2606Ter)	DMD	X	31679429	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.8358G>A (p.Trp2786Ter)	DMD	X	31507313	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.8464C>T (p.Gln2822Ter)	DMD	X	31496871	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.8668G>A (p.Glu2890Lys)	DMD	X	31478983	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.9346C>T (p.Gln3116Ter)	DMD	X	31223062	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.961-5831C>T	DMD	X	32650983	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.9851G>A (p.Trp3284Ter)	DMD	X	31182861	Dilated cardiomyopathy 3B
NM_004006.2(DMD):c.2302C>T (p.Arg768Ter)	DMD	X	32501833	Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.1952G>A (p.Trp651Ter)	DMD	X	32565742	Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.3121C>T (p.Gln1041Ter)	DMD	X	32468539	Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.6292C>T (p.Arg2098Ter)	DMD	X	32217062	Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.4375C>T (p.Arg1459Ter)	DMD	X	32389644	Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.8443C>T (p.Gln2815Ter)	DMD	X	31496892	Dilated cardiomyopathy 3B;Duchenne muscular dystrophy
NM_004006.2(DMD):c.10141C>T (p.Arg3381Ter)	DMD	X	31178751	Dilated cardiomyopathy 3B;Duchenne muscular dystrophy;not provided
NM_000941.2(POR):c.1706G>A (p.Cys569Tyr)	POR	7	75985959	Disordered steroidogenesis due to cytochrome p450 oxidoreductase deficiency
NM_000416.2(IFNGR1):c.200+1G>A	IFNGR1	6	137206962	Disseminated atypical mycobacterial infection

NM_000416.2(IFNGR1):c.230G>A (p.Cys77Tyr)	IFNGR1	6	137206279	Disseminated atypical mycobacterial infection
NM_002470.3(MYH3):c.1123G>A (p.Glu375Lys)	MYH3	17	10645725	Distal arthrogryposis type 2B
NM_002470.3(MYH3):c.533C>T (p.Thr178Ile)	MYH3	17	10650374	Distal arthrogryposis type 2B;Freeman-Sheldon syndrome
NM_003282.3(TNNI2):c.521G>A (p.Arg174Gln)	TNNI2	11	1841523	Distal arthrogryposis type 2B;not provided
NM_003289.3(TPM2):c.397C>T (p.Arg133Trp)	TPM2	9	35685529	Distal arthrogryposis type 2B;not provided
NM_002470.3(MYH3):c.700G>A (p.Ala234Thr)	MYH3	17	10648592	Distal arthrogryposis type 2B;not provided
NM_001540.3(HSPB1):c.452C>T (p.Thr151Ile)	HSPB1	7	76304007	Distal hereditary motor neuronopathy type 2B
NM_001540.3(HSPB1):c.545C>T (p.Pro182Leu)	HSPB1	7	76304100	Distal hereditary motor neuronopathy type 2B
NM_001540.3(HSPB1):c.544C>T (p.Pro182Ser)	HSPB1	7	76304099	Distal hereditary motor neuronopathy type 2B
NM_004082.4(DCTN1):c.175G>A (p.Gly59Ser)	DCTN1	2	74378104	Distal hereditary motor neuronopathy type 7B
NM_002180.2(IGHMBP2):c.127C>T (p.Arg43Ter)	IGHMBP2	11	68906109	Distal hereditary motor neuronopathy;Spinal muscular atrophy, distal, autosomal recessive, 1;not provided
NM_133378.4(TTN):c.100186C>T (p.Gln33396Ter)	TTN	2	178527098	Distal myopathy Markesbery-Griggs type
NM_133378.4(TTN):c.12064C>T (p.Arg4022Ter)	TTN	2	178733497	Distal myopathy Markesbery-Griggs type
NM_133378.4(TTN):c.5047C>T (p.Arg1683Ter)	TTN	2	178776817	Distal myopathy Markesbery-Griggs type
NM_001256850.1(TTN):c.100831C>T (p.Arg33611Ter)	TTN	2	178530861	Distal myopathy Markesbery-Griggs type;Familial hypertrophic cardiomyopathy 9;Hereditary myopathy with early respiratory failure;Limb-girdle muscular dystrophy, type 2J;Myopathy, early-onset, with fatal cardiomyopathy;not provided
NM_033337.2(CAV3):c.137C>T (p.Ala46Val)	CAV3	3	8745548	Distal myopathy, Tateyama type;Rippling muscle disease 2;not provided
NM_000052.6(ATP7A):c.2981C>T (p.Thr994Ile)	ATP7A	X	78029314	Distal spinal muscular atrophy, X-linked 3
NM_000052.6(ATP7A):c.4156C>T (p.Pro1386Ser)	ATP7A	X	78045502	Distal spinal muscular atrophy, X-linked 3
NM_005251.2(FOXC2):c.374C>T (p.Ser125Leu)	FOXC2	16	86567709	Distichiasis-lymphedema syndrome
NM_005251.2(FOXC2):c.362G>A (p.Arg121His)	FOXC2	16	86567697	Distichiasis-lymphedema syndrome
NM_000234.2(LIG1):c.1696G>A (p.Glu566Lys)	LIG1	19	48133011	DNA ligase I deficiency
NM_000234.2(LIG1):c.2311C>T (p.Arg771Trp)	LIG1	19	48121244	DNA ligase I deficiency
NM_003286.3(TOP1):c.1252G>A (p.Glu418Lys)	TOP1	20	41101297	DNA topoisomerase I, camptothecin-resistant
NM_001067.3(TOP2A):c.1460G>A (p.Arg487Lys)	TOP2A	17	40408007	Dna topoisomerase II, resistance to inhibition of, by amsacrine
NM_017780.3(CHD7):c.6892C>T (p.Gln2298Ter)	CHD7	8	60854479	Dolichocephaly;Hypothyroidism;Myopia;Poor coordination;Progressive hearing impairment;Scoliosis
NM_000094.3(COL7A1):c.5443G>A (p.Gly1815Arg)	COL7A1	3	48578497	Dominant dystrophic epidermolysis bullosa with absence of skin;Nail disorder, nonsyndromic congenital, 8;Recessive dystrophic epidermolysis bullosa
NM_000094.3(COL7A1):c.6007G>A (p.Gly2003Arg)	COL7A1	3	48575512	Dominant dystrophic epidermolysis bullosa with absence of skin;not provided
NM_015560.2(OPA1):c.899G>A (p.Gly300Glu)	OPA1	3	193637980	Dominant hereditary optic atrophy
NM_015560.2(OPA1):c.985-1G>A	OPA1	3	193642764	Dominant hereditary optic atrophy
NM_130837.2(OPA1):c.740G>A (p.Arg247His)	OPA1	3	193626153	Dominant hereditary optic atrophy
NM_015560.2(OPA1):c.1202G>A (p.Gly401Asp)	OPA1	3	193643434	Dominant hereditary optic atrophy
NM_015560.2(OPA1):c.1140+1G>A	OPA1	3	193643050	Dominant hereditary optic atrophy
NM_015560.2(OPA1):c.869G>A (p.Arg290Gln)	OPA1	3	193637280	Dominant hereditary optic atrophy;not provided
NM_015560.2(OPA1):c.1096C>T (p.Arg366Ter)	OPA1	3	193643005	Dominant hereditary optic atrophy;not provided
NM_015560.2(OPA1):c.1669C>T (p.Arg557Ter)	OPA1	3	193647144	Dominant hereditary optic atrophy;not provided
NM_015560.2(OPA1):c.2257C>T (p.Gln753Ter)	OPA1	3	193658977	Dominant hereditary optic atrophy;not provided
NM_015560.2(OPA1):c.870+1G>A	OPA1	3	193637282	Dominant hereditary optic atrophy;not provided
NM_015560.2(OPA1):c.2569C>T (p.Arg857Ter)	OPA1	3	193664952	Dominant hereditary optic atrophy;not provided
NM_004525.2(LRP2):c.2640-1G>A	LRP2	2	169256237	Donnai Barrow syndrome
NM_004525.2(LRP2):c.8452+1G>A	LRP2	2	169201627	Donnai Barrow syndrome
NM_004525.2(LRP2):c.10195C>T (p.Arg3399Ter)	LRP2	2	169178001	Donnai Barrow syndrome

NM_004525.2(LRP2):c.1093C>T (p.Arg365Ter)	LRP2	2	169282951	Donnai Barrow syndrome	
NM_004525.2(LRP2):c.6160G>A (p.Asp2054Asn)	LRP2	2	169212088	Donnai Barrow syndrome	
NM_000787.3(DBH):c.301G>A (p.Val101Met)	DBH	9	133636672	Dopamine beta hydroxylase deficiency	
NM_000787.3(DBH):c.1033G>A (p.Asp345Asn)	DBH	9	133647854	Dopamine beta hydroxylase deficiency	
NM_001492.5(GDF1):c.800G>A (p.Cys267Tyr)	GDF1	19	18868916	Double outlet right ventricle	
NM_152305.2(POGLUT1):c.11G>A (p.Trp4Ter)	POGLUT1	3	119469032	Dowling-degos disease 4	
NM_152305.2(POGLUT1):c.652C>T (p.Arg218Ter)	POGLUT1	3	119486846	Dowling-degos disease 4	
NM_152305.2(POGLUT1):c.835C>T (p.Arg279Trp)	POGLUT1	3	119490588	Dowling-degos disease 4	
NM_004992.3(MECP2):c.419C>T (p.Ala140Val)	MECP2	X	154031409	Downslanted palpebral fissures;Hearing impairment;Intellectual disability;Mental retardation, X-linked, syndromic 13;Microcephaly;Micrognathia;Motor delay;Postnatal growth retardation;Rett syndrome;Stenosis of the external auditory canal;not provided	
NM_001039348.2(EFEMP1):c.1033C>T (p.Arg345Trp)	EFEMP1	2	55871091	Doyne honeycomb retinal dystrophy;Malattia leventinese	
NM_024426.4(WT1):c.1193G>A (p.Cys398Tyr)	WT1	11	32396313	Drash syndrome	
NM_024426.4(WT1):c.1432+5G>A	WT1	11	32391967	Drash syndrome;Frasier syndrome;Hereditary nephrotic syndrome	
NM_024426.4(WT1):c.1372C>T (p.Arg458Ter)	WT1	11	32392032	Drash syndrome;Frasier syndrome;Wilms tumor 1	
NM_024426.4(WT1):c.1301G>A (p.Arg434His)	WT1	11	32392704	Drash syndrome;not provided	
NM_024426.4(WT1):c.1288C>T (p.Arg430Ter)	WT1	11	32392717	Drash syndrome;Wilms tumor 1	
NM_001822.5(CHN1):c.668C>T (p.Ala223Val)	CHN1	2	174824478	Duane syndrome type 2	
NM_001822.5(CHN1):c.682G>A (p.Gly228Ser)	CHN1	2	174824464	Duane syndrome type 2	
NM_001822.5(CHN1):c.937G>A (p.Glu313Lys)	CHN1	2	174811538	Duane syndrome type 2	
NM_001822.5(CHN1):c.422C>T (p.Pro141Leu)	CHN1	2	174877967	Duane syndrome type 2	
NM_001822.5(CHN1):c.754C>T (p.Pro252Ser)	CHN1	2	174812441	Duane syndrome type 2	
NM_020436.4(SALL4):c.1954C>T (p.Gln652Ter)	SALL4	20	51790529	Duane-radial ray syndrome	
NM_020436.4(SALL4):c.2593C>T (p.Arg865Ter)	SALL4	20	51789010	Duane-radial ray syndrome	
NM_020436.4(SALL4):c.1849C>T (p.Arg617Ter)	SALL4	20	51790634	Duane-radial ray syndrome	
NM_020436.4(SALL4):c.2713C>T (p.Arg905Ter)	SALL4	20	51788890	Duane-radial ray syndrome	
NM_000392.4(ABCC2):c.2302C>T (p.Arg768Trp)	ABCC2	10	99818820	Dubin-Johnson syndrome	
NM_000392.4(ABCC2):c.3449G>A (p.Arg1150His)	ABCC2	10	99836125	Dubin-Johnson syndrome	
NM_000392.4(ABCC2):c.3196C>T (p.Arg1066Ter)	ABCC2	10	99832069	Dubin-Johnson syndrome	
NM_000392.4(ABCC2):c.1967+1G>A	ABCC2	10	99811603	Dubin-Johnson syndrome	
NM_000392.4(ABCC2):c.3741+1G>A	ABCC2	10	99842094	Dubin-Johnson syndrome	
NM_004006.2(DMD):c.5551C>T (p.Gln1851Ter)	DMD	X	32345978	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.6955C>T (p.Gln2319Ter)	DMD	X	31875331	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.253C>T (p.Gln85Ter)	DMD	X	32844794	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.2017C>T (p.Gln673Ter)	DMD	X	32545310	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.178C>T (p.Gln60Ter)	DMD	X	32849736	Duchenne muscular dystrophy	
NM_000109.3(DMD):c.700C>T (p.Gln234Ter)	DMD	X	32699219	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1489C>T (p.Gln497Ter)	DMD	X	32595870	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.3188G>A (p.Trp1063Ter)	DMD	X	32464674	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.4213C>T (p.Gln1405Ter)	DMD	X	32411772	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.4414C>T (p.Gln1472Ter)	DMD	X	32389605	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.6790C>T (p.Gln2264Ter)	DMD	X	31929718	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.8357G>A (p.Trp2786Ter)	DMD	X	31507314	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.7817G>A (p.Trp2606Ter)	DMD	X	31679430	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.7755G>A (p.Trp2585Ter)	DMD	X	31679492	Duchenne muscular dystrophy	

NM_004006.2(DMD):c.5917C>T (p.Gln1973Ter)	DMD	X	32342105	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.5641C>T (p.Gln1881Ter)	DMD	X	32343232	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.5131C>T (p.Gln1711Ter)	DMD	X	32364605	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.4240C>T (p.Gln1414Ter)	DMD	X	32390175	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.3427C>T (p.Gln1143Ter)	DMD	X	32463444	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.2797C>T (p.Gln933Ter)	DMD	X	32484925	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.2407C>T (p.Gln803Ter)	DMD	X	32491492	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.2368C>T (p.Gln790Ter)	DMD	X	32501767	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1683G>A (p.Trp561Ter)	DMD	X	32573766	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1663C>T (p.Gln555Ter)	DMD	X	32573786	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1388G>A (p.Trp463Ter)	DMD	X	32614397	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1331+1G>A	DMD	X	32644131	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1324C>T (p.Gln442Ter)	DMD	X	32644139	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.355C>T (p.Gln119Ter)	DMD	X	32823297	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.94-1G>A	DMD	X	32849821	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.5506C>T (p.Gln1836Ter)	DMD	X	32346023	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.4870C>T (p.Gln1624Ter)	DMD	X	32365175	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1504C>T (p.Gln502Ter)	DMD	X	32595855	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.5032C>T (p.Gln1678Ter)	DMD	X	32364704	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.457C>T (p.Gln153Ter)	DMD	X	32816541	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1594C>T (p.Gln532Ter)	DMD	X	32595765	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1150-1G>A	DMD	X	32644314	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.6223C>T (p.Gln2075Ter)	DMD	X	32287596	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.3747G>A (p.Trp1249Ter)	DMD	X	32448495	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.2861G>A (p.Trp954Ter)	DMD	X	32472252	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.9563+1G>A	DMD	X	31209497	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.4483C>T (p.Gln1495Ter)	DMD	X	32389536	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.4312C>T (p.Gln1438Ter)	DMD	X	32390103	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.8209C>T (p.Gln2737Ter)	DMD	X	31627681	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.4071+1G>A	DMD	X	32438240	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.2665C>T (p.Arg889Ter)	DMD	X	32485057	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.2202G>A (p.Trp734Ter)	DMD	X	32518098	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.2077C>T (p.Gln693Ter)	DMD	X	32545250	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1653G>A (p.Trp551Ter)	DMD	X	32573796	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.1061G>A (p.Trp354Ter)	DMD	X	32645052	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.8914C>T (p.Gln2972Ter)	DMD	X	31478129	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.6118-1G>A	DMD	X	32287702	Duchenne muscular dystrophy	
NM_004006.2(DMD):c.10019G>A (p.Cys3340Tyr)	DMD	X	31180437	Duchenne muscular dystrophy, mental retardation, and absence of erg b-wave	
NM_004006.2(DMD):c.4729C>T (p.Arg1577Ter)	DMD	X	32380626	Duchenne muscular dystrophy;not provided	
NM_001142548.1(RAD54L):c.973G>A (p.Gly325Arg)	RAD54L	1	46267540	Ductal breast carcinoma	
NM_005689.3(ABCB6):c.1736G>A (p.Gly579Glu)	ABCB6	2	219213310	Dyschromatosis universalis hereditaria 3	
NM_002049.3(GATA1):c.613G>A (p.Val205Met)	GATA1	X	48792337	Dyserythropoietic anemia with thrombocytopenia;GATA-1-related thrombocytopenia with dyserythropoiesis	
NM_000508.3(FGA):c.103C>T (p.Arg35Cys)	FGA	4	154589514	Dysfibrinogenemia	
NM_000508.4(FGA):c.104G>A (p.Arg35His)	FGA	4	154589513	Dysfibrinogenemia	

NM_003494.3(DYSF):c.1663C>T (p.Arg555Trp)	DYSF	2	71551631	DYSF-Related Disorders;Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_198253.2(TERT):c.2045G>A (p.Gly682Asp)	TERT	5	1279376	Dyskeratosis congenita autosomal dominant
NR_001566.1(TERC):n.143G>A	TERC	3	169764918	Dyskeratosis congenita autosomal dominant
NR_001566.1(TERC):n.35C>T	TERC	3	169765026	Dyskeratosis congenita autosomal dominant
NM_001099274.1(TINF2):c.844C>T (p.Arg282Cys)	TINF2	14	24240636	Dyskeratosis congenita autosomal dominant;Dyskeratosis congenita, autosomal dominant, 3;not provided
NM_198253.2(TERT):c.2110C>T (p.Pro704Ser)	TERT	5	1279311	Dyskeratosis congenita autosomal dominant;Dyskeratosis congenita, autosomal recessive, 4
NM_001099274.1(TINF2):c.847C>T (p.Pro283Ser)	TINF2	14	24240633	Dyskeratosis congenita autosomal dominant;not provided
NR_001566.1(TERC):n.116C>T	TERC	3	169764945	Dyskeratosis congenita autosomal dominant;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 2
NM_018648.3(NOP10):c.100C>T (p.Arg34Trp)	NOP10	15	34342063	Dyskeratosis congenita autosomal recessive 1
NM_017838.3(NHP2):c.376G>A (p.Val126Met)	NHP2	5	178149799	Dyskeratosis congenita autosomal recessive 1;Dyskeratosis congenita, autosomal recessive 2
NM_198253.2(TERT):c.2431C>T (p.Arg811Cys)	TERT	5	1271156	Dyskeratosis congenita autosomal recessive 1;Dyskeratosis congenita, autosomal recessive, 4
NM_198253.2(TERT):c.2701C>T (p.Arg901Trp)	TERT	5	1264546	Dyskeratosis congenita autosomal recessive 1;Dyskeratosis congenita, autosomal recessive, 4
NM_001363.4(DKC1):c.1205G>A (p.Gly402Glu)	DKC1	X	154774651	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.1050G>A (p.Met350Ile)	DKC1	X	154773144	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.1075G>A (p.Asp359Asn)	DKC1	X	154773169	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.1150C>T (p.Pro384Ser)	DKC1	X	154773244	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.1151C>T (p.Pro384Leu)	DKC1	X	154773245	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.1156G>A (p.Ala386Thr)	DKC1	X	154774602	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.1204G>A (p.Gly402Arg)	DKC1	X	154774650	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.121G>A (p.Glu41Lys)	DKC1	X	154765480	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.1226C>T (p.Pro409Leu)	DKC1	X	154774672	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.200C>T (p.Thr67Ile)	DKC1	X	154765935	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.29C>T (p.Pro10Leu)	DKC1	X	154764911	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.5C>T (p.Ala2Val)	DKC1	X	154762970	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.911G>A (p.Ser304Asn)	DKC1	X	154769306	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.949C>T (p.Leu317Phe)	DKC1	X	154770792	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.965G>A (p.Arg322Gln)	DKC1	X	154770808	Dyskeratosis congenita X-linked
NM_001363.4(DKC1):c.146C>T (p.Thr49Met)	DKC1	X	154765505	Dyskeratosis congenita X-linked;Hoyeraal Hreidarsson syndrome;not provided
NM_001363.4(DKC1):c.1223C>T (p.Thr408Ile)	DKC1	X	154774669	Dyskeratosis congenita X-linked;not provided
NM_198253.2(TERT):c.2098C>T (p.Gln700Ter)	TERT	5	1279323	Dyskeratosis congenita, autosomal dominant, 2
NM_198253.2(TERT):c.1892G>A (p.Arg631Gln)	TERT	5	1280216	Dyskeratosis congenita, autosomal dominant, 2;Idiopathic fibrosing alveolitis, chronic form
NM_001099274.1(TINF2):c.805C>T (p.Gln269Ter)	TINF2	14	24240675	Dyskeratosis congenita, autosomal dominant, 3
NM_001099274.1(TINF2):c.811C>T (p.Gln271Ter)	TINF2	14	24240669	Dyskeratosis congenita, autosomal dominant, 3
NM_001099274.1(TINF2):c.845G>A (p.Arg282His)	TINF2	14	24240635	Dyskeratosis congenita, autosomal dominant, 3;Revesz syndrome
NM_032957.4(RTEL1):c.1933G>A (p.Ala645Thr)	RTEL1	20	63689115	Dyskeratosis congenita, autosomal dominant, 4
NM_032957.4(RTEL1):c.3028C>T (p.Arg1010Ter)	RTEL1	20	63693247	Dyskeratosis congenita, autosomal dominant, 4;Dyskeratosis congenita, autosomal recessive, 5;not provided
NM_002582.3(PARN):c.1045C>T (p.Arg349Trp)	PARN	16	14584383	Dyskeratosis congenita, autosomal recessive 6
NM_018081.2(WRAP53):c.1192C>T (p.Arg398Trp)	WRAP53	17	7702770	Dyskeratosis congenita, autosomal recessive, 3
NM_018081.2(WRAP53):c.1126C>T (p.His376Tyr)	WRAP53	17	7702514	Dyskeratosis congenita, autosomal recessive, 3
NM_018081.2(WRAP53):c.1303G>A (p.Gly435Arg)	WRAP53	17	7703027	Dyskeratosis congenita, autosomal recessive, 3
NM_032957.4(RTEL1):c.2941C>T (p.Arg981Trp)	RTEL1	20	63693160	Dyskeratosis congenita, autosomal recessive, 5

NM_032957.4(RTEL1):c.823G>A (p.Glu275Lys)	RTEL1	20	63672607	Dyskeratosis congenita, autosomal recessive, 5
NM_016434.3(RTEL1):c.2233G>A (p.Val745Met)	RTEL1	20	63690178	Dyskeratosis congenita, autosomal recessive, 5
NM_016434.3(RTEL1):c.2141+5G>A	RTEL1	20	63689870	Dyskeratosis congenita, autosomal recessive, 5
NM_016434.3(RTEL1):c.2920C>T (p.Arg974Ter)	RTEL1	20	63693211	Dyskeratosis congenita, autosomal recessive, 5;Idiopathic fibrosing alveolitis, chronic form
RTEL1:c.2402G>A (p.Arg801His)	RTEL1	20	63695619	Dyskeratosis congenita, autosomal recessive, 5;Idiopathic fibrosing alveolitis, chronic form;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3
NM_001363.4(DKC1):c.1058C>T (p.Ala353Val)	DKC1	X	154773152	Dyskeratosis congenita;Dyskeratosis congenita X-linked
NM_002582.3(PARN):c.1148C>T (p.Ala383Val)	PARN	16	14582225	Dyskeratosis congenita;Dyskeratosis congenita, autosomal recessive 6
NM_183357.2(ADCY5):c.2088+1G>A	ADCY5	3	123325321	Dyskinesia, familial, with facial myokymia
NM_183357.2(ADCY5):c.1253G>A (p.Arg418Gln)	ADCY5	3	123352463	Dyskinesia, familial, with facial myokymia
NM_183357.2(ADCY5):c.2176G>A (p.Ala726Thr)	ADCY5	3	123319754	Dyskinesia, familial, with facial myokymia;not provided
NM_183357.2(ADCY5):c.1252C>T (p.Arg418Trp)	ADCY5	3	123352464	Dyskinesia, familial, with facial myokymia;not provided
NM_000301.3(PLG):c.1858G>A (p.Ala620Thr)	PLG	6	160738593	Dysplasminogenemia
NM_000301.3(PLG):c.2251G>A (p.Gly751Arg)	PLG	6	160752240	Dysplasminogenemia
NM_145239.2(PRRT2):c.796C>T (p.Arg266Trp)	PRRT2	16	29813850	Dystonia 10
NM_145239.2(PRRT2):c.487C>T (p.Gln163Ter)	PRRT2	16	29813541	Dystonia 10;Infantile convulsions and paroxysmal choreoathetosis, familial
NM_145239.2(PRRT2):c.748C>T (p.Gln250Ter)	PRRT2	16	29813802	Dystonia 10;Seizures, benign familial infantile, 2
NM_152296.4(ATP1A3):c.1838C>T (p.Thr613Met)	ATP1A3	19	41978041	Dystonia 12
NM_152296.4(ATP1A3):c.829G>A (p.Glu277Lys)	ATP1A3	19	41985082	Dystonia 12
NM_152296.4(ATP1A3):c.2051C>T (p.Ser684Phe)	ATP1A3	19	41976459	Dystonia 12
NM_152296.4(ATP1A3):c.2267G>A (p.Arg756His)	ATP1A3	19	41970539	Dystonia 12
NM_152296.4(ATP1A3):c.946G>A (p.Gly316Ser)	ATP1A3	19	41984965	Dystonia 12
NM_003690.4(PRKRA):c.665C>T (p.Pro222Leu)	PRKRA	2	178436264	Dystonia 16
NM_002143.2(HPCA):c.568G>A (p.Ala190Thr)	HPCA	1	32893848	Dystonia 2, torsion, autosomal recessive
NM_000718.3(CACNA1B):c.4166G>A (p.Arg1389His)	CACNA1B	9	138058108	Dystonia 23
NM_001142339.2(GNAL):c.409G>A (p.Val137Met)	GNAL	18	11824933	Dystonia 25
NM_001142339.2(GNAL):c.463G>A (p.Glu155Lys)	GNAL	18	11824987	Dystonia 25
NM_001142339.2(GNAL):c.61C>T (p.Arg21Ter)	GNAL	18	11752494	Dystonia 25
NM_001282684.1(KCTD17):c.434G>A (p.Arg145His)	KCTD17	22	37057420	Dystonia 26, myoclonic
NM_004369.3(COL6A3):c.7660G>A (p.Ala2554Thr)	COL6A3	2	237344358	Dystonia 27
NM_014727.2(KMT2B):c.1633C>T (p.Arg545Ter)	KMT2B	19	35720980	DYSTONIA 28, CHILDHOOD-ONSET
NM_014727.2(KMT2B):c.2428C>T (p.Gln810Ter)	KMT2B	19	35721775	DYSTONIA 28, CHILDHOOD-ONSET
NM_014727.2(KMT2B):c.1690C>T (p.Arg564Ter)	KMT2B	19	35721037	DYSTONIA 28, CHILDHOOD-ONSET
NM_014727.2(KMT2B):c.7549C>T (p.Arg2517Trp)	KMT2B	19	35737262	DYSTONIA 28, CHILDHOOD-ONSET
NR_104387.1(TAF1):n.5894C>T	TAF1	X	71529785	Dystonia 3, torsion, X-linked
NM_000161.2(GCH1):c.262C>T (p.Arg88Trp)	GCH1	14	54902402	Dystonia 5, Dopa-responsive type
NM_000161.2(GCH1):c.602G>A (p.Gly201Glu)	GCH1	14	54845792	Dystonia 5, Dopa-responsive type
NM_000161.2(GCH1):c.142C>T (p.Gln48Ter)	GCH1	14	54902522	Dystonia 5, Dopa-responsive type
NM_006516.2(SLC2A1):c.376C>T (p.Arg126Cys)	SLC2A1	1	42930766	Dystonia 9;Epilepsy, idiopathic generalized, susceptibility to, 12;GLUT1 deficiency syndrome 1;GLUT1 deficiency syndrome 2;not provided
NM_006516.2(SLC2A1):c.634C>T (p.Arg212Cys)	SLC2A1	1	42929918	Dystonia 9;not provided
NM_016011.4(MECR):c.695G>A (p.Gly232Glu)	MECR	1	29202004	DYSTONIA, CHILDHOOD-ONSET, WITH OPTIC ATROPHY AND BASAL GANGLIA ABNORMALITIES
NM_000161.2(GCH1):c.323G>A (p.Gly108Asp)	GCH1	14	54902341	Dystonia, dopa-responsive, with or without hyperphenylalaninemia, autosomal recessive
NM_199292.2(TH):c.1481C>T (p.Thr494Met)	TH	11	2164339	Dystonia;Segawa syndrome, autosomal recessive
NM_000094.3(COL7A1):c.5797C>T (p.Arg1933Ter)	COL7A1	3	48576272	Dystrophic epidermolysis bullosa

NM_000094.3(COL7A1):c.1637-1G>A	COL7A1	3	48590817	Dystrophic epidermolysis bullosa;not provided
NM_003165.3(STXBP1):c.1434G>A (p.Trp478Ter)	STXBP1	9	127678505	Early infantile epileptic encephalopathy
NM_007254.3(PNKP):c.526C>T (p.Leu176Phe)	PNKP	19	49864376	Early infantile epileptic encephalopathy 10
NM_007254.3(PNKP):c.976G>A (p.Glu326Lys)	PNKP	19	49862424	Early infantile epileptic encephalopathy 10;not provided
NM_001040142.1(SCN2A):c.304C>T (p.Arg102Ter)	SCN2A	2	165297053	Early infantile epileptic encephalopathy 11
NM_001040142.1(SCN2A):c.4726G>A (p.Gly1576Arg)	SCN2A	2	165386920	Early infantile epileptic encephalopathy 11
NM_021007.2(SCN2A):c.2558G>A (p.Arg853Gln)	SCN2A	2	165342465	Early infantile epileptic encephalopathy 11;not provided
NM_014191.3(SCN8A):c.4850G>A (p.Arg1617Gln)	SCN8A	12	51806336	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.4351G>A (p.Gly1451Ser)	SCN8A	12	51789350	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.2300C>T (p.Thr767Ile)	SCN8A	12	51751523	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.641G>A (p.Gly214Asp)	SCN8A	12	51688784	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.1588C>T (p.Arg530Trp)	SCN8A	12	51706668	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.2668G>A (p.Ala890Thr)	SCN8A	12	51765794	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.4873G>A (p.Gly1625Arg)	SCN8A	12	51806359	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.5615G>A (p.Arg1872Gln)	SCN8A	12	51807101	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.2003C>T (p.Thr668Ile)	SCN8A	12	51745907	Early infantile epileptic encephalopathy 13
NM_014191.3(SCN8A):c.4948G>A (p.Ala1650Thr)	SCN8A	12	51806434	Early infantile epileptic encephalopathy 13;Epileptic encephalopathy;not provided
NM_014191.3(SCN8A):c.2549G>A (p.Arg850Gln)	SCN8A	12	51765575	Early infantile epileptic encephalopathy 13;not provided
NM_020822.2(KCNT1):c.2800G>A (p.Ala934Thr)	KCNT1	9	135779429	Early infantile epileptic encephalopathy 14
NM_020822.2(KCNT1):c.1283G>A (p.Arg428Gln)	KCNT1	9	135765706	Early infantile epileptic encephalopathy 14;Epilepsy, nocturnal frontal lobe, 5;not provided
NM_020822.2(KCNT1):c.1421G>A (p.Arg474His)	KCNT1	9	135768848	Early infantile epileptic encephalopathy 14;not provided
NM_020822.2(KCNT1):c.862G>A (p.Gly288Ser)	KCNT1	9	135759686	Early infantile epileptic encephalopathy 14;not provided
NM_015284.3(SZT2):c.73C>T (p.Arg25Ter)	SZT2	1	43403222	Early infantile epileptic encephalopathy 18
NM_015284.3(SZT2):c.2092C>T (p.Gln698Ter)	SZT2	1	43423153	Early infantile epileptic encephalopathy 18
NM_003159.2(CDKL5):c.863C>T (p.Thr288Ile)	CDKL5	X	18598499	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.1039C>T (p.Gln347Ter)	CDKL5	X	18603963	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.2413C>T (p.Gln805Ter)	CDKL5	X	18625164	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.2593C>T (p.Gln865Ter)	CDKL5	X	18628467	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.539C>T (p.Pro180Leu)	CDKL5	X	18584338	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.146-1G>A	CDKL5	X	18575353	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.622C>T (p.Gln208Ter)	CDKL5	X	18588021	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.404-1G>A	CDKL5	X	18581890	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.2494C>T (p.Gln832Ter)	CDKL5	X	18625245	Early infantile epileptic encephalopathy 2
NM_001323289.1(CDKL5):c.2716C>T (p.Gln906Ter)	CDKL5	X	18628590	Early infantile epileptic encephalopathy 2
NM_003159.2(CDKL5):c.587C>T (p.Ser196Leu)	CDKL5	X	18587986	Early infantile epileptic encephalopathy 2;Epileptic encephalopathy;not provided
NM_003159.2(CDKL5):c.533G>A (p.Arg178Gln)	CDKL5	X	18584332	Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.1954C>T (p.Gln652Ter)	CDKL5	X	18608820	Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.2152G>A (p.Val718Met)	CDKL5	X	18609570	Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.400C>T (p.Arg134Ter)	CDKL5	X	18579965	Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.2376+1G>A	CDKL5	X	18619967	Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.99+5G>A	CDKL5	X	18510859	Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.2596C>T (p.Gln866Ter)	CDKL5	X	18628470	Early infantile epileptic encephalopathy 2;not provided
NM_003159.2(CDKL5):c.199C>T (p.Leu67Phe)	CDKL5	X	18575407	Early infantile epileptic encephalopathy 2;Rett syndrome
NM_015509.3(NECAP1):c.142C>T (p.Arg48Ter)	NECAP1	12	8089982	Early infantile epileptic encephalopathy 21
NM_001134771.1(SLC12A5):c.1652G>A (p.Gly551Asp)	SLC12A5	20	46045891	Early infantile epileptic encephalopathy 34

NM_003165.3(STXBP1):c.1631G>A (p.Gly544Asp)	STXBP1	9	127682489	Early infantile epileptic encephalopathy 4
NM_003165.3(STXBP1):c.539G>A (p.Cys180Tyr)	STXBP1	9	127663314	Early infantile epileptic encephalopathy 4
NM_003165.3(STXBP1):c.169+1G>A	STXBP1	9	127653797	Early infantile epileptic encephalopathy 4
NM_003165.3(STXBP1):c.902+1G>A	STXBP1	9	127668188	Early infantile epileptic encephalopathy 4
NM_003165.3(STXBP1):c.1006C>T (p.Gln336Ter)	STXBP1	9	127672093	Early infantile epileptic encephalopathy 4
NM_003165.3(STXBP1):c.1162C>T (p.Arg388Ter)	STXBP1	9	127675855	Early infantile epileptic encephalopathy 4;not provided
NM_003165.3(STXBP1):c.847G>A (p.Glu283Lys)	STXBP1	9	127668132	Early infantile epileptic encephalopathy 4;not provided
NM_003165.3(STXBP1):c.364C>T (p.Arg122Ter)	STXBP1	9	127661140	Early infantile epileptic encephalopathy 4;not provided
NM_003165.3(STXBP1):c.416C>T (p.Pro139Leu)	STXBP1	9	127661192	Early infantile epileptic encephalopathy 4;not provided
NM_003165.3(STXBP1):c.568C>T (p.Arg190Trp)	STXBP1	9	127663343	Early infantile epileptic encephalopathy 4;not provided
NM_003165.3(STXBP1):c.703C>T (p.Arg235Ter)	STXBP1	9	127666205	Early infantile epileptic encephalopathy 4;not provided
NM_003165.3(STXBP1):c.875G>A (p.Arg292His)	STXBP1	9	127668160	Early infantile epileptic encephalopathy 4;not provided
NM_003165.3(STXBP1):c.1651C>T (p.Arg551Cys)	STXBP1	9	127682509	Early infantile epileptic encephalopathy 4;not provided
NM_172107.3(KCNQ2):c.628C>T (p.Arg210Cys)	KCNQ2	20	63444721	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.2147C>T (p.Thr716Ile)	KCNQ2	20	63407116	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.917C>T (p.Ala306Val)	KCNQ2	20	63439608	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.388G>A (p.Glu130Lys)	KCNQ2	20	63445364	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.1956G>A (p.Pro652=)	KCNQ2	20	63407307	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.1641G>A (p.Arg547=)	KCNQ2	20	63413572	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.1053C>T (p.Leu351=)	KCNQ2	20	63433874	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.926C>T (p.Ala309Val)	KCNQ2	20	63439599	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.757G>A (p.Ala253Thr)	KCNQ2	20	63442465	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.643G>A (p.Gly215Arg)	KCNQ2	20	63444706	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.471G>A (p.Trp157Ter)	KCNQ2	20	63445281	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.341C>T (p.Thr114Ile)	KCNQ2	20	63446793	Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.901G>A (p.Gly301Ser)	KCNQ2	20	63439624	Early infantile epileptic encephalopathy 7;Epileptic encephalopathy
NM_172107.3(KCNQ2):c.802C>T (p.Leu268Phe)	KCNQ2	20	63442420	Early infantile epileptic encephalopathy 7;Epileptic encephalopathy
NM_172107.3(KCNQ2):c.997C>T (p.Arg333Trp)	KCNQ2	20	63438651	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.638G>A (p.Arg213Gln)	KCNQ2	20	63444711	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.794C>T (p.Ala265Val)	KCNQ2	20	63442428	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.1687G>A (p.Asp563Asn)	KCNQ2	20	63413526	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.1682C>T (p.Pro561Leu)	KCNQ2	20	63413531	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.1058G>A (p.Arg353His)	KCNQ2	20	63433869	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.601C>T (p.Arg201Cys)	KCNQ2	20	63444748	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.431G>A (p.Arg144Gln)	KCNQ2	20	63445321	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.629G>A (p.Arg210His)	KCNQ2	20	63444720	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.827C>T (p.Thr276Ile)	KCNQ2	20	63439698	Early infantile epileptic encephalopathy 7;not provided
NM_172107.3(KCNQ2):c.620G>A (p.Arg207Gln)	KCNQ2	20	63444729	Early infantile epileptic encephalopathy 7;Seizures, benign familial neonatal, 1, and/or myokymia;not provided
NM_172107.3(KCNQ2):c.602G>A (p.Arg201His)	KCNQ2	20	63444747	Early infantile epileptic encephalopathy 7;Severe intellectual deficiency
NM_001173479.1(ARHGEF9):c.4C>T (p.Gln2Ter)	ARHGEF9	X	63785142	Early infantile epileptic encephalopathy 8
NM_001184880.1(PCDH19):c.253C>T (p.Gln85Ter)	PCDH19	X	100408345	Early infantile epileptic encephalopathy 9
NM_001184880.1(PCDH19):c.814C>T (p.Gln272Ter)	PCDH19	X	100407784	Early infantile epileptic encephalopathy 9
NM_001184880.1(PCDH19):c.490C>T (p.Gln164Ter)	PCDH19	X	100408108	Early infantile epileptic encephalopathy 9

NM_014191.3(SCN8A):c.5614C>T (p.Arg1872Trp)	SCN8A	12	51807100	Early infantile epileptic encephalopathy;Early infantile epileptic encephalopathy 13;Epileptic encephalopathy;not provided
NM_020988.2(GNAO1):c.607G>A (p.Gly203Arg)	GNAO1	16	56336744	Early infantile epileptic encephalopathy;Early infantile epileptic encephalopathy 17;not provided
NM_003165.3(STXBP1):c.1439C>T (p.Pro480Leu)	STXBP1	9	127678510	Early infantile epileptic encephalopathy;Early infantile epileptic encephalopathy 4;not provided
NM_003165.3(STXBP1):c.1217G>A (p.Arg406His)	STXBP1	9	127675910	Early infantile epileptic encephalopathy;Early infantile epileptic encephalopathy 4;not provided
NM_172107.3(KCNQ2):c.868G>A (p.Gly290Ser)	KCNQ2	20	63439657	Early infantile epileptic encephalopathy;Early infantile epileptic encephalopathy 7
NM_172107.3(KCNQ2):c.881C>T (p.Ala294Val)	KCNQ2	20	63439644	Early infantile epileptic encephalopathy;Early infantile epileptic encephalopathy 7;Epileptic encephalopathy;not provided
NM_172107.3(KCNQ2):c.1678C>T (p.Arg560Trp)	KCNQ2	20	63413535	Early infantile epileptic encephalopathy;Early infantile epileptic encephalopathy 7;not provided
NM_006920.4(SCN1A):c.4874G>A (p.Arg1625Gln)	SCN1A	2	165992368	Early infantile epileptic encephalopathy;Epileptic encephalopathy Lennox-Gastaut type;Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy
NM_003165.3(STXBP1):c.1099C>T (p.Arg367Ter)	STXBP1	9	127673250	Early infantile epileptic encephalopathy;Epileptic encephalopathy;not provided
NM_020988.2(GNAO1):c.625C>T (p.Arg209Cys)	GNAO1	16	56336762	Early infantile epileptic encephalopathy;not provided
NM_001165963.1(SCN1A):c.2792G>A (p.Arg931His)	SCN1A	2	166037930	Early infantile epileptic encephalopathy;Severe myoclonic epilepsy in infancy;not provided
NM_001191061.1(SLC25A22):c.617C>T (p.Pro206Leu)	SLC25A22	11	792429	Early myoclonic encephalopathy
NM_024698.5(SLC25A22):c.418C>T (p.Gln140Ter)	SLC25A22	11	792722	Early myoclonic encephalopathy
NM_175629.2(DNMT3A):c.1204C>T (p.Gln402Ter)	DNMT3A	2	25246695	Early T cell progenitor acute lymphoblastic leukemia
NM_001005463.2(EBF3):c.487C>T (p.Arg163Trp)	EBF3	10	129957325	EBF3-related disorder
NM_022336.3(EDAR):c.1072C>T (p.Arg358Ter)	EDAR	2	108897182	Ectodermal dysplasia 10a, hypohidrotic/hair/tooth type, autosomal dominant;not provided
NM_145861.2(EDARADD):c.120+1G>A	EDARADD	1	236409275	Ectodermal dysplasia 11b, hypohidrotic/hair/tooth type, autosomal recessive
NM_000299.3(PKP1):c.910C>T (p.Gln304Ter)	PKP1	1	201317635	Ectodermal dysplasia skin fragility syndrome
NM_020529.2(NFKBIA):c.32G>A (p.Trp11Ter)	NFKBIA	14	35404613	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, autosomal dominant
NM_030916.2(NECTIN4):c.851G>A (p.Arg284Gln)	NECTIN4	1	161076355	Ectodermal dysplasia-syndactyly syndrome 1
NM_030916.2(NECTIN4):c.554C>T (p.Thr185Met)	NECTIN4	1	161077629	Ectodermal dysplasia-syndactyly syndrome 1
NM_019032.5(ADAMTSL4):c.79-1G>A	ADAMTSL4	1	150552897	Ectopia lentis, isolated autosomal recessive
NM_019032.5(ADAMTSL4):c.2008C>T (p.Arg670Ter)	ADAMTSL4	1	150557296	Ectopia lentis, isolated autosomal recessive
NM_000138.4(FBN1):c.2920C>T (p.Arg974Cys)	FBN1	15	48490013	Ectopia lentis, isolated, autosomal dominant;Marfan syndrome
NM_000138.4(FBN1):c.7339G>A (p.Glu2447Lys)	FBN1	15	48425483	Ectopia lentis, isolated, autosomal dominant;Marfan syndrome;not provided
NM_000138.4(FBN1):c.718C>T (p.Arg240Cys)	FBN1	15	48537629	Ectopia lentis, isolated, autosomal dominant;Marfan syndrome;not provided
NM_003722.4(TP63):c.728G>A (p.Arg243Gln)	TP63	3	189864380	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3
NM_003722.4(TP63):c.727C>T (p.Arg243Trp)	TP63	3	189864379	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3;not provided
NM_003722.4(TP63):c.1028G>A (p.Arg343Gln)	TP63	3	189868615	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3;not provided
NM_003722.4(TP63):c.953G>A (p.Arg318His)	TP63	3	189867903	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3;Rapp-Hodgkin ectodermal dysplasia syndrome;not provided
NM_138615.2(DHX30):c.2344C>T (p.Arg782Trp)	DHX30	3	47848237	EEG abnormality;Failure to thrive;Generalized hypotonia;Global developmental delay;Hirsutism;Intellectual disability;Microcephaly;Short stature
NM_007255.2(B4GALT7):c.641G>A (p.Cys214Tyr)	B4GALT7	5	177608540	Ehlers-Danlos syndrome progeroid type
NM_007255.2(B4GALT7):c.808C>T (p.Arg270Cys)	B4GALT7	5	177608994	Ehlers-Danlos syndrome progeroid type;Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects;not provided
NM_007255.2(B4GALT7):c.421C>T (p.Arg141Trp)	B4GALT7	5	177607309	Ehlers-Danlos syndrome progeroid type;not provided
NM_000089.3(COL1A2):c.1404+1G>A	COL1A2	7	94412122	Ehlers-Danlos syndrome, autosomal recessive, cardiac valvular form
NM_000093.4(COL5A1):c.4466G>A (p.Gly1489Glu)	COL5A1	9	134820135	Ehlers-Danlos syndrome, classic type
NM_000093.4(COL5A1):c.2374C>T (p.Arg792Ter)	COL5A1	9	134774901	Ehlers-Danlos syndrome, classic type
NM_001278074.1(COL5A1):c.2034+1G>A	COL5A1	9	134763738	Ehlers-Danlos syndrome, classic type
NM_000093.4(COL5A1):c.2430+1G>A	COL5A1	9	134780147	Ehlers-Danlos syndrome, classic type

NM_000093.4(COL5A1):c.3184C>T (p.Arg1062Ter)	COL5A1	9	134805044	Ehlers-Danlos syndrome, classic type; not provided
NM_000302.3(PLOD1):c.955C>T (p.Arg319Ter)	PLOD1	1	11958627	Ehlers-Danlos syndrome, hydroxylysine-deficient
NM_000302.3(PLOD1):c.2032G>A (p.Gly678Arg)	PLOD1	1	11974656	Ehlers-Danlos syndrome, hydroxylysine-deficient
NM_000302.3(PLOD1):c.579+1G>A	PLOD1	1	11952736	Ehlers-Danlos syndrome, hydroxylysine-deficient
NM_000302.3(PLOD1):c.2008C>T (p.Arg670Ter)	PLOD1	1	11972977	Ehlers-Danlos syndrome, hydroxylysine-deficient
NM_130468.3(CHST14):c.842C>T (p.Pro281Leu)	CHST14	15	40472055	Ehlers-Danlos syndrome, musculocontractural type
NM_001080976.2(DSE):c.803C>T (p.Ser268Leu)	DSE	6	116431086	Ehlers-Danlos syndrome, musculocontractural type 2
NM_080605.3(B3GALT6):c.16C>T (p.Arg6Trp)	B3GALT6	1	1232294	Ehlers-Danlos syndrome, progeroid type, 2
NM_000090.3(COL3A1):c.2437G>A (p.Gly813Ser)	COL3A1	2	189002343	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3149G>A (p.Gly1050Asp)	COL3A1	2	189006400	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1149+1G>A (p.Gly351_Pro383del)	COL3A1	2	188993460	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3093+1G>A	COL3A1	2	189006260	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.746G>A (p.Gly249Asp)	COL3A1	2	188990308	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1655G>A (p.Gly552Glu)	COL3A1	2	188996171	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2931+1G>A (p.Gly942_Lys977del)	COL3A1	2	189004365	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3041G>A (p.Gly1014Glu)	COL3A1	2	189006207	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3518G>A (p.Gly1173Glu)	COL3A1	2	189008135	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3563G>A (p.Gly1188Glu)	COL3A1	2	189008961	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1869+5G>A	COL3A1	2	188997394	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1997G>A (p.Gly666Asp)	COL3A1	2	188998693	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3302G>A (p.Gly1101Glu)	COL3A1	2	189007546	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2212G>A (p.Gly738Ser)	COL3A1	2	188999560	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2600G>A (p.Gly867Asp)	COL3A1	2	189003457	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2276G>A (p.Gly759Asp)	COL3A1	2	188999888	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1033G>A (p.Gly345Arg)	COL3A1	2	188992923	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.548G>A (p.Gly183Asp)	COL3A1	2	188988100	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2222G>A (p.Gly741Asp)	COL3A1	2	188999570	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1916G>A (p.Gly639Glu)	COL3A1	2	188997746	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.951+5G>A (p.Gly300_Ala317del)	COL3A1	2	188991727	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2823+1G>A (p.Gly942Valfs*28)	COL3A1	2	189004144	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3491G>A (p.Gly1164Glu)	COL3A1	2	189008108	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2780G>A (p.Gly927Asp)	COL3A1	2	189004100	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2861G>A (p.Gly954Glu)	COL3A1	2	189004294	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2140G>A (p.Gly714Arg)	COL3A1	2	188999488	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2824G>A (p.Gly942Arg)	COL3A1	2	189004257	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3301G>A (p.Gly1101Arg)	COL3A1	2	189007545	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.907G>A (p.Gly303Arg)	COL3A1	2	188991678	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.556G>A (p.Gly186Ser)	COL3A1	2	188988108	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2842G>A (p.Gly948Arg)	COL3A1	2	189004275	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3417+5G>A (p.Gly1122_Arg1139del)	COL3A1	2	189007943	Ehlers-Danlos syndrome, type 4

NM_000090.3(COL3A1):c.2771G>A (p.Gly924Asp)	COL3A1	2	189004091	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.899G>A (p.Gly300Asp)	COL3A1	2	188991670	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1988G>A (p.Gly663Asp)	COL3A1	2	188998684	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2941G>A (p.Gly981Arg)	COL3A1	2	189005359	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2978G>A (p.Gly993Asp)	COL3A1	2	189005396	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2114G>A (p.Gly705Glu)	COL3A1	2	188999376	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2337G>A (p.Lys779=)	COL3A1	2	189001450	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.798+1G>A (p.Phe268Leufs*10)	COL3A1	2	188990361	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3509G>A (p.Gly1170Asp)	COL3A1	2	189008126	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2564G>A (p.Gly855Asp)	COL3A1	2	189003421	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3847C>T (p.Gln1283Ter)	COL3A1	2	189010201	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3239G>A (p.Gly1080Asp)	COL3A1	2	189006974	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3473G>A (p.Gly1158Asp)	COL3A1	2	189008090	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2068G>A (p.Gly690Arg)	COL3A1	2	188999330	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1466G>A (p.Gly489Glu)	COL3A1	2	188995056	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.809G>A (p.Gly270Glu)	COL3A1	2	188991014	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3070C>T (p.Arg1024Ter)	COL3A1	2	189006236	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3255+1G>A (p.Gly1068_Pro1085del)	COL3A1	2	189006991	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3508G>A (p.Gly1170Ser)	COL3A1	2	189008125	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2203G>A (p.Gly735Arg)	COL3A1	2	188999551	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1330G>A (p.Gly444Arg)	COL3A1	2	188994577	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1979G>A (p.Gly660Asp)	COL3A1	2	188998675	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2555G>A (p.Gly852Asp)	COL3A1	2	189003412	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3437G>A (p.Gly1146Glu)	COL3A1	2	189008054	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1087G>A (p.Gly363Ser)	COL3A1	2	188993397	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1475G>A (p.Gly492Glu)	COL3A1	2	188995065	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3255+5G>A (p.Gly1068_Pro1085del)	COL3A1	2	189006995	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3562G>A (p.Gly1188Arg)	COL3A1	2	189008960	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2708G>A (p.Gly903Glu)	COL3A1	2	189004028	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3039+1G>A (p.Asp1013_Gly1014insMSSSFYSTSQ)	COL3A1	2	189005458	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1761+5G>A (p.Gly555_Asp587del)	COL3A1	2	188996501	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2888G>A (p.Gly963Asp)	COL3A1	2	189004321	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2105G>A (p.Gly702Asp)	COL3A1	2	188999367	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.996+1G>A (p.Gly318_Pro332del)	COL3A1	2	188992229	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2825G>A (p.Gly942Glu)	COL3A1	2	189004258	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2554-1G>A (p.Gly852Valfs*384)	COL3A1	2	189003410	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3517G>A (p.Gly1173Arg)	COL3A1	2	189008134	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1501G>A (p.Gly501Arg)	COL3A1	2	188995091	Ehlers-Danlos syndrome, type 4	

NM_000090.3(COL3A1):c.2510G>A (p.Gly837Asp)	COL3A1	2	189003019	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1808G>A (p.Gly603Asp)	COL3A1	2	188997211	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.844G>A (p.Gly282Arg)	COL3A1	2	188991049	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3391G>A (p.Gly1131Ser)	COL3A1	2	189007912	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1149+5G>A (p.Gly351_Pro383del)	COL3A1	2	188993464	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3167G>A (p.Gly1056Asp)	COL3A1	2	189006418	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2185G>A (p.Gly729Arg)	COL3A1	2	188999533	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1142G>A (p.Gly381Asp)	COL3A1	2	188993452	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3140G>A (p.Gly1047Asp)	COL3A1	2	189006391	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2150G>A (p.Gly717Asp)	COL3A1	2	188999498	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3032G>A (p.Gly1011Glu)	COL3A1	2	189005450	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3220G>A (p.Gly1074Ser)	COL3A1	2	189006955	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.683G>A (p.Gly228Glu)	COL3A1	2	188989442	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.637G>A (p.Gly213Ser)	COL3A1	2	188989396	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.755G>A (p.Gly252Asp)	COL3A1	2	188990317	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2915G>A (p.Gly972Asp)	COL3A1	2	189004348	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3319G>A (p.Gly1107Arg)	COL3A1	2	189007563	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3193G>A (p.Gly1065Arg)	COL3A1	2	189006444	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.4254G>A (p.Thr1418=)	COL3A1	2	189010890	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2833G>A (p.Gly945Ser)	COL3A1	2	189004266	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3039+5G>A (p.Asp1013_Gly1014insVNSSFYSTSQ)	COL3A1	2	189005462	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1124G>A (p.Gly375Glu)	COL3A1	2	188993434	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.737G>A (p.Gly246Glu)	COL3A1	2	188990142	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1923+1G>A (p.Gly624_Gln641del)	COL3A1	2	188997754	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2987G>A (p.Gly996Glu)	COL3A1	2	189005405	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2905G>A (p.Gly969Arg)	COL3A1	2	189004338	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2168G>A (p.Gly723Asp)	COL3A1	2	188999516	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1194+1G>A (p.Gly384_Met398del)	COL3A1	2	188994083	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.4294C>T (p.Arg1432Ter)	COL3A1	2	189011667	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2087G>A (p.Gly696Asp)	COL3A1	2	188999349	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.836G>A (p.Gly279Asp)	COL3A1	2	188991041	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2933G>A (p.Gly978Asp)	COL3A1	2	189005351	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.647G>A (p.Gly216Glu)	COL3A1	2	188989406	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2699G>A (p.Gly900Asp)	COL3A1	2	189004019	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.592G>A (p.Gly198Arg)	COL3A1	2	188988599	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2770G>A (p.Gly924Ser)	COL3A1	2	189004090	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.791G>A (p.Gly264Glu)	COL3A1	2	188990353	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2194G>A (p.Gly732Arg)	COL3A1	2	188999542	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3284G>A (p.Gly1095Asp)	COL3A1	2	189007528	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1898G>A (p.Gly633Glu)	COL3A1	2	188997728	Ehlers-Danlos syndrome, type 4

NM_000090.3(COL3A1):c.2824-1G>A (p.Gly942Glufs*294)	COL3A1	2	189004256	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1961G>A (p.Gly654Glu)	COL3A1	2	188998303	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1358G>A (p.Gly453Asp)	COL3A1	2	188994734	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1862G>A (p.Gly621Glu)	COL3A1	2	188997382	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2645G>A (p.Gly882Asp)	COL3A1	2	189003771	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2113G>A (p.Gly705Arg)	COL3A1	2	188999375	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.611G>A (p.Gly204Asp)	COL3A1	2	188988618	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2293G>A (p.Gly765Ser)	COL3A1	2	189001406	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.656G>A (p.Gly219Asp)	COL3A1	2	188989415	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1267G>A (p.Gly423Ser)	COL3A1	2	188994306	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.926G>A (p.Gly309Glu)	COL3A1	2	188991697	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1384G>A (p.Gly462Ser)	COL3A1	2	188994760	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1844G>A (p.Gly615Glu)	COL3A1	2	188997364	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.782G>A (p.Gly261Asp)	COL3A1	2	188990344	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2445+5G>A (p.Gly798_Pro815del)	COL3A1	2	189002356	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1249G>A (p.Gly417Arg)	COL3A1	2	188994288	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2176G>A (p.Gly726Arg)	COL3A1	2	188999524	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.593G>A (p.Gly198Glu)	COL3A1	2	188988600	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2501G>A (p.Gly834Asp)	COL3A1	2	189003010	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2022G>A (p.Lys674=)	COL3A1	2	188998718	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2285G>A (p.Gly762Asp)	COL3A1	2	189001398	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.827G>A (p.Gly276Asp)	COL3A1	2	188991032	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3545G>A (p.Gly1182Glu)	COL3A1	2	189008943	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3496C>T (p.Arg1166Ter)	COL3A1	2	189008113	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3419G>A (p.Gly1140Glu)	COL3A1	2	189008036	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2995G>A (p.Gly999Ser)	COL3A1	2	189005413	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.951+1G>A (p.Gly300_Ala317del)	COL3A1	2	188991723	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.665G>A (p.Gly222Asp)	COL3A1	2	188989424	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2870G>A (p.Gly957Asp)	COL3A1	2	189004303	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.701G>A (p.Gly234Asp)	COL3A1	2	188990106	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3490G>A (p.Gly1164Arg)	COL3A1	2	189008107	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.3023G>A (p.Gly1008Asp)	COL3A1	2	189005441	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.690+1G>A (p.Gly213_Asp230del)	COL3A1	2	188989450	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2753G>A (p.Gly918Glu)	COL3A1	2	189004073	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2607+5G>A (p.Gly852_Pro869del)	COL3A1	2	189003469	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2337+1G>A (p.Gly762_Lys779del)	COL3A1	2	189001451	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.2553+1G>A (p.Gly816_Ala851del)	COL3A1	2	189003063	Ehlers-Danlos syndrome, type 4	
NM_000090.3(COL3A1):c.1915G>A (p.Gly639Arg)	COL3A1	2	188997745	Ehlers-Danlos syndrome, type 4	

NM_000090.3(COL3A1):c.582+5G>A (p.Gly177_Pro194del)	COL3A1	2	188988139	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.764G>A (p.Gly255Glu)	COL3A1	2	188990326	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3325C>T (p.Arg1109Ter)	COL3A1	2	189007569	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.728G>A (p.Gly243Glu)	COL3A1	2	188990133	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3266G>A (p.Gly1089Asp)	COL3A1	2	189007510	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.998G>A (p.Gly333Asp)	COL3A1	2	188992888	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2860G>A (p.Gly954Arg)	COL3A1	2	189004293	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.636+1G>A (p.Gly195_Ser212del)	COL3A1	2	188988644	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.30G>A (p.Trp10Ter)	COL3A1	2	188974519	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2141G>A (p.Gly714Glu)	COL3A1	2	188999489	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1646G>A (p.Gly549Glu)	COL3A1	2	188996162	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2186G>A (p.Gly729Glu)	COL3A1	2	188999534	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3544G>A (p.Gly1182Arg)	COL3A1	2	189008942	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2221G>A (p.Gly741Ser)	COL3A1	2	188999569	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.636+5G>A (p.Gly195_Ser212del)	COL3A1	2	188988648	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2402G>A (p.Gly801Asp)	COL3A1	2	189002308	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1763G>A (p.Gly588Asp)	COL3A1	2	188997166	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1258G>A (p.Gly420Ser)	COL3A1	2	188994297	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1556G>A (p.Gly519Glu)	COL3A1	2	188995738	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2131G>A (p.Gly711Ser)	COL3A1	2	188999479	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1096G>A (p.Gly366Arg)	COL3A1	2	188993406	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2177G>A (p.Gly726Glu)	COL3A1	2	188999525	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.1970G>A (p.Gly657Glu)	COL3A1	2	188998312	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3572G>A (p.Gly1191Asp)	COL3A1	2	189008970	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2815G>A (p.Gly939Ser)	COL3A1	2	189004135	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2023-1G>A (p.Gly675Valfs*116)	COL3A1	2	188999284	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.610G>A (p.Gly204Ser)	COL3A1	2	188988617	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.539G>A (p.Gly180Asp)	COL3A1	2	188988091	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2735G>A (p.Gly912Asp)	COL3A1	2	189004055	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3482G>A (p.Gly1161Glu)	COL3A1	2	189008099	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.799G>A (p.Gly267Ser)	COL3A1	2	188991004	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3410G>A (p.Gly1137Asp)	COL3A1	2	189007931	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2914G>A (p.Gly972Ser)	COL3A1	2	189004347	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.3833G>A (p.Trp1278Ter)	COL3A1	2	189010187	Ehlers-Danlos syndrome, type 4
NM_000090.3(COL3A1):c.2356G>A (p.Gly786Arg)	COL3A1	2	189001554	Ehlers-Danlos syndrome, type 4;Ehlers-Danlos syndrome, type 4 variant
NM_000090.3(COL3A1):c.1347+1G>A (p.Arg449_Gly450insValSerPheThrAlaThrAspLeu+)	COL3A1	2	188994595	Ehlers-Danlos syndrome, type 4;Ehlers-Danlos syndrome, type 4 variant
NM_000090.3(COL3A1):c.3554G>A (p.Gly1185Asp)	COL3A1	2	189008952	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.1744G>A (p.Gly582Ser)	COL3A1	2	188996479	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.547G>A (p.Gly183Ser)	COL3A1	2	188988099	Ehlers-Danlos syndrome, type 4;not provided

NM_000090.3(COL3A1):c.3417+1G>A (p.Gly1122_Arg1139del+)	COL3A1	2	189007939	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.1618G>A (p.Gly540Arg)	COL3A1	2	188996134	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.1268G>A (p.Gly423Asp)	COL3A1	2	188994307	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.976C>T (p.Arg326Ter)	COL3A1	2	188992208	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.709G>A (p.Gly237Arg)	COL3A1	2	188990114	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.3356G>A (p.Gly1119Asp)	COL3A1	2	189007600	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.962G>A (p.Gly321Asp)	COL3A1	2	188992194	Ehlers-Danlos syndrome, type 4;not provided
NM_000090.3(COL3A1):c.970G>A (p.Gly324Ser)	COL3A1	2	188992202	Ehlers-Danlos syndrome, type 4;Thoracic aortic aneurysm and aortic dissection
NM_000090.3(COL3A1):c.2096G>A (p.Gly699Asp)	COL3A1	2	188999358	Ehlers-Danlos syndrome, type 4;Thoracic aortic aneurysm and aortic dissection
NM_000090.3(COL3A1):c.1662+1G>A (p.Gly537_Pro554del)	COL3A1	2	188996179	Ehlers-Danlos syndrome, type 4;Thoracic aortic aneurysm and aortic dissection;not provided
NG_007400.1:g.8207G>A	COL1A1	17	50198433	Ehlers-Danlos syndrome, type 7A
NM_001733.4(C1R):c.890G>A (p.Gly297Asp)	C1R	12	7088865	Ehlers-Danlos syndrome, type 8
NM_014244.4(ADAMTS2):c.673C>T (p.Gln225Ter)	ADAMTS2	5	179272926	Ehlers-Danlos syndrome, type vii, autosomal recessive
NM_014244.4(ADAMTS2):c.2384G>A (p.Trp795Ter)	ADAMTS2	5	179130005	Ehlers-Danlos syndrome, type vii, autosomal recessive
NM_019105.6(TNXB):c.12214C>T (p.Arg4072Cys)	TNXB	6	32042353	Ehlers-Danlos-like syndrome due to tenascin-X deficiency
NM_020451.2(SELENON):c.818G>A (p.Gly273Glu)	SELENON	1	25809096	Eichsfeld type congenital muscular dystrophy
NM_020451.2(SEPN1):c.1385G>A (p.Sec462=)	SELENON	1	25812790	Eichsfeld type congenital muscular dystrophy
NM_020451.2(SELENON):c.1397G>A (p.Arg466Gln)	SELENON	1	25813890	Eichsfeld type congenital muscular dystrophy;not provided
NM_020451.2(SELENON):c.1315C>T (p.Arg439Ter)	SELENON	1	25812720	Eichsfeld type congenital muscular dystrophy;not provided
NM_000316.2(PTH1R):c.1453C>T (p.Arg485Ter)	PTH1R	3	46903327	Eiken skeletal dysplasia
NM_004006.2(DMD):c.6139C>T (p.Gln2047Ter)	DMD	X	32287680	Elevated serum creatine phosphokinase
NM_003126.2(SPTA1):c.145C>T (p.Leu49Phe)	SPTA1	1	158685227	Elliptocytosis 2
NM_003126.2(SPTA1):c.121C>T (p.Arg41Trp)	SPTA1	1	158685251	Elliptocytosis 2
NM_003126.2(SPTA1):c.2465_2587del123 (p.Lys823_Gly863del)	SPTA1	1	158661410	Elliptocytosis 2
NM_003126.2(SPTA1):c.82C>T (p.Arg28Cys)	SPTA1	1	158685290	Elliptocytosis 2;Hereditary pyropoikilocytosis
NM_003126.2(SPTA1):c.83G>A (p.Arg28His)	SPTA1	1	158685289	Elliptocytosis 2;Hereditary pyropoikilocytosis;not provided
NM_000117.2(EMD):c.3G>A (p.Met1Ile)	EMD	X	154379487	Emery-Dreifuss muscular dystrophy 1, X-linked
NM_033071.3(SYNE1):c.24071G>A (p.Arg8024His)	SYNE1	6	152151987	Emery-Dreifuss muscular dystrophy 4, autosomal dominant
NM_033071.3(SYNE1):c.11908C>T (p.Arg3970Ter)	SYNE1	6	152344185	Emery-Dreifuss muscular dystrophy 4, autosomal dominant
NM_033071.3(SYNE1):c.21721C>T (p.Gln7241Ter)	SYNE1	6	152219113	Emery-Dreifuss muscular dystrophy 4, autosomal dominant;Spinocerebellar ataxia, autosomal recessive 8
NM_024334.2(TMEM43):c.253G>A (p.Glu85Lys)	TMEM43	3	14130912	Emery-Dreifuss muscular dystrophy 7, autosomal dominant
NM_000117.2(EMD):c.130C>T (p.Gln44Ter)	EMD	X	154379737	Emery-Dreifuss muscular dystrophy, X-linked;not provided
NM_017565.3(FAM20A):c.406C>T (p.Arg136Ter)	FAM20A	17	68555742	Enamel-renal syndrome
NM_017565.3(FAM20A):c.826C>T (p.Arg276Ter)	FAM20A	17	68542796	Enamel-renal syndrome
NM_017565.3(FAM20A):c.1432C>T (p.Arg478Ter)	FAM20A	17	68537671	Enamel-renal syndrome
m.5545C>T	MT-TW	MT	5545	Encephalocardiomyopathy, mitochondrial
NM_001277061.1(MFF):c.892C>T (p.Arg298Ter)	MFF	2	227355756	ENCEPHALOPATHY DUE TO DEFECTIVE MITOCHONDRIAL AND PEROXISOMAL FISSION 2
NM_020194.5(MFF):c.190C>T (p.Gln64Ter)	MFF	2	227330777	ENCEPHALOPATHY DUE TO DEFECTIVE MITOCHONDRIAL AND PEROXISOMAL FISSION 2;Global developmental delay;Mitochondrial encephalomyopathy
NM_006267.4(RANBP2):c.1754C>T (p.Thr585Met)	RANBP2	2	108751993	Encephalopathy, acute, infection-induced, 3, suceptibility to
NM_006267.4(RANBP2):c.1958C>T (p.Thr653Ile)	RANBP2	2	108753466	Encephalopathy, acute, infection-induced, 3, suceptibility to

NM_001122752.1(SERPINI1):c.1175G>A (p.Gly392Glu)	SERPINI1	3	167825265	Encephalopathy, familial, with neuroserpin inclusion bodies
NM_005025.4(SERPINI1):c.1174G>A (p.Gly392Arg)	SERPINI1	3	167825264	Encephalopathy, familial, with neuroserpin inclusion bodies
NM_012062.4(DNM1L):c.1085G>A (p.Gly362Asp)	DNM1L	12	32731019	Encephalopathy, lethal, due to defective mitochondrial and peroxisomal fission
NM_012062.4(DNM1L):c.1084G>A (p.Gly362Ser)	DNM1L	12	32731018	Encephalopathy, lethal, due to defective mitochondrial and peroxisomal fission
NM_012062.4(DNM1L):c.1048G>A (p.Gly350Arg)	DNM1L	12	32722602	Encephalopathy, lethal, due to defective mitochondrial and peroxisomal fission
NM_012062.4(DNM1L):c.1207C>T (p.Arg403Cys)	DNM1L	12	32731362	Encephalopathy, lethal, due to defective mitochondrial and peroxisomal fission;not provided
m.5549G>A	MT-TW	MT	5549	Encephalopathy, mitochondrial
m.586G>A	MT-TF	MT	586	Encephalopathy, mitochondrial
NM_005993.4(TBCD):c.2314C>T (p.Arg772Cys)	TBCD	17	82924992	ENCEPHALOPATHY, PROGRESSIVE, EARLY-ONSET, WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM
NM_005993.4(TBCD):c.2761G>A (p.Ala921Thr)	TBCD	17	82929180	ENCEPHALOPATHY, PROGRESSIVE, EARLY-ONSET, WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM
NM_005993.4(TBCD):c.3365C>T (p.Pro1122Leu)	TBCD	17	82938132	ENCEPHALOPATHY, PROGRESSIVE, EARLY-ONSET, WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM
NM_005993.4(TBCD):c.1876G>A (p.Ala626Thr)	TBCD	17	82906007	ENCEPHALOPATHY, PROGRESSIVE, EARLY-ONSET, WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM
NM_005993.4(TBCD):c.1130G>A (p.Arg377Gln)	TBCD	17	82807650	ENCEPHALOPATHY, PROGRESSIVE, EARLY-ONSET, WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM
NM_144772.2(NAXE):c.196C>T (p.Gln66Ter)	NAXE	1	156592114	ENCEPHALOPATHY, PROGRESSIVE, EARLY-ONSET, WITH BRAIN EDEMA AND/OR LEUKOENCEPHALOPATHY
NM_144772.2(NAXE):c.516+1G>A	NAXE	1	156592671	ENCEPHALOPATHY, PROGRESSIVE, EARLY-ONSET, WITH BRAIN EDEMA AND/OR LEUKOENCEPHALOPATHY
NM_032504.1(UNC80):c.565G>A (p.Val189Met)	UNC80	2	209777524	Encephalopathy;Hypotonia, infantile, with psychomotor retardation and characteristic facies 2
NM_032504.1(UNC80):c.1078C>T (p.Arg360Ter)	UNC80	2	209813719	Encephalopathy;Hypotonia, infantile, with psychomotor retardation and characteristic facies 2
NM_032504.1(UNC80):c.3793C>T (p.Arg1265Ter)	UNC80	2	209872917	Encephalopathy;Hypotonia, infantile, with psychomotor retardation and characteristic facies 2
NM_016513.4(ICK):c.815G>A (p.Arg272Gln)	ICK	6	53016099	Endocrine-cerebroosteodysplasia
NM_000179.2(MSH6):c.2731C>T (p.Arg911Ter)	MSH6	2	47800714	Endometrial carcinoma;Hereditary cancer-predisposing syndrome;Hereditary nonpolyposis colorectal cancer type 5;Lynch syndrome;not provided
NM_004985.4(KRAS):c.40G>A (p.Val14Ile)	KRAS	12	25245345	Endometrial carcinoma;Noonan syndrome 3;Rasopathy;not provided
NM_000314.6(PTEN):c.253+1G>A	PTEN	10	87931090	Endometrial carcinoma;not provided
NM_018082.5(POLR3B):c.2774C>T (p.Pro925Leu)	POLR3B	12	106496115	Endosteal sclerosis-cerebellar hypoplasia syndrome
NM_005677.3(COLQ):c.943C>T (p.Arg315Ter)	COLQ	3	15458197	Endplate acetylcholinesterase deficiency
NM_014249.3(NR2E3):c.226C>T (p.Arg76Trp)	NR2E3	15	71811590	Enhanced s-cone syndrome
NM_006177.3(NRL):c.91C>T (p.Arg31Ter)	NRL	14	24082758	Enhanced s-cone syndrome
NM_014249.3(NR2E3):c.932G>A (p.Arg311Gln)	NR2E3	15	71813573	Enhanced s-cone syndrome;Goldmann-Favre syndrome;NR2E3-Related Disorders;not provided
NM_000441.1(SLC26A4):c.1115C>T (p.Ala372Val)	SLC26A4	7	107689166	Enlarged vestibular aqueduct syndrome
NM_000441.1(SLC26A4):c.1489G>A (p.Gly497Ser)	SLC26A4	7	107695984	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.2162C>T (p.Thr721Met)	SLC26A4	7	107710126	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.578C>T (p.Thr193Ile)	SLC26A4	7	107674326	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_012188.4(FOXI1):c.800G>A (p.Arg267Gln)	FOXI1	5	170108274	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.1336C>T (p.Gln446Ter)	SLC26A4	7	107694475	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.2015G>A (p.Gly672Glu)	SLC26A4	7	107702038	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.2188C>T (p.Gln730Ter)	SLC26A4	7	107710152	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.2089+1G>A	SLC26A4	7	107704386	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.2215C>T (p.Gln739Ter)	SLC26A4	7	107710179	Enlarged vestibular aqueduct syndrome;Pendred's syndrome

NM_000441.1(SLC26A4):c.1554G>A (p.Trp518Ter)	SLC26A4	7	107698051	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.1544+1G>A	SLC26A4	7	107696040	Enlarged vestibular aqueduct syndrome;Pendred's syndrome
NM_000441.1(SLC26A4):c.1226G>A (p.Arg409His)	SLC26A4	7	107690200	Enlarged vestibular aqueduct syndrome;Pendred's syndrome;not provided
NM_000441.1(SLC26A4):c.1229C>T (p.Thr410Met)	SLC26A4	7	107690203	Enlarged vestibular aqueduct syndrome;Pendred's syndrome;not provided
NM_000441.1(SLC26A4):c.1614+1G>A	SLC26A4	7	107698112	Enlarged vestibular aqueduct syndrome;Pendred's syndrome;not provided
NM_000441.1(SLC26A4):c.1001+1G>A	SLC26A4	7	107683538	Enlarged vestibular aqueduct syndrome;Pendred's syndrome;SLC26A4-Related Disorders;not provided
NM_002772.2(TMPRSS15):c.2569C>T (p.Arg857Ter)	TMPRSS15	21	18281139	Enterokinase deficiency
NM_002772.2(TMPRSS15):c.781C>T (p.Gln261Ter)	TMPRSS15	21	18359856	Enterokinase deficiency
NM_002524.4(NRAS):c.101C>T (p.Pro34Leu)	NRAS	1	114716060	Epidermal nevus;Noonan syndrome 1
NM_007267.7(TMC6):c.280C>T (p.Arg94Ter)	TMC6	17	78125876	EPIDERMODYSPLASIA VERRUCIFORMIS
NM_000445.4(PLEC):c.6169C>T (p.Gln2057Ter)	PLEC	8	143923841	Epidermolysa bullosa simplex and limb girdle muscular dystrophy
NM_000445.4(PLEC):c.6955C>T (p.Arg2319Ter)	PLEC	8	143923055	Epidermolysa bullosa simplex and limb girdle muscular dystrophy
NM_201383.2(PLEC):c.4267C>T (p.Gln1423Ter)	PLEC	8	143925674	Epidermolysa bullosa simplex and limb girdle muscular dystrophy
NM_201383.2(PLEC):c.7192C>T (p.Arg2398Ter)	PLEC	8	143922749	Epidermolysa bullosa simplex and limb girdle muscular dystrophy
NM_000094.3(COL7A1):c.8479C>T (p.Gln2827Ter)	COL7A1	3	48565458	Epidermolysis bullosa dystrophica inversa, autosomal recessive
NM_000526.4(KRT14):c.373C>T (p.Arg125Cys)	KRT14	17	41586462	Epidermolysis bullosa herpetiformis, Dowling-Meara;not provided
NM_000526.4(KRT14):c.374G>A (p.Arg125His)	KRT14	17	41586461	Epidermolysis bullosa herpetiformis, Dowling-Meara;not provided
NM_000526.4(KRT14):c.1264G>A (p.Glu422Lys)	KRT14	17	41583245	Epidermolysis bullosa herpetiformis, Dowling-Meara;not provided
NM_000424.3(KRT5):c.523C>T (p.Leu175Phe)	KRT5	12	52519774	Epidermolysis bullosa herpetiformis, Dowling-Meara;not provided
NM_000424.3(KRT5):c.1429G>A (p.Glu477Lys)	KRT5	12	52516647	Epidermolysis bullosa herpetiformis, Dowling-Meara;not provided
NM_000213.4(ITGB4):c.1660C>T (p.Arg554Ter)	ITGB4	17	75736053	Epidermolysis bullosa junctionalis with pyloric atresia
NM_001005619.1(ITGB4):c.4592G>A (p.Trp1531Ter)	ITGB4	17	75755785	Epidermolysis bullosa junctionalis with pyloric atresia
NM_000213.4(ITGB4):c.3674G>A (p.Arg1225His)	ITGB4	17	75750992	Epidermolysis bullosa junctionalis with pyloric atresia
NM_000445.4(PLEC):c.1486C>T (p.Gln496Ter)	PLEC	8	143933210	Epidermolysis bullosa junctionalis with pyloric atresia
NM_000213.4(ITGB4):c.182G>A (p.Cys61Tyr)	ITGB4	17	75727423	Epidermolysis bullosa junctionalis with pyloric atresia;not provided
NM_000213.4(ITGB4):c.3793+1G>A	ITGB4	17	75751112	Epidermolysis bullosa junctionalis with pyloric atresia;not provided
NM_000213.4(ITGB4):c.3841C>T (p.Arg1281Trp)	ITGB4	17	75752221	Epidermolysis bullosa junctionalis with pyloric atresia;not provided
NM_000094.3(COL7A1):c.6724G>A (p.Gly2242Arg)	COL7A1	3	48573047	Epidermolysis bullosa pruriginosa, autosomal dominant
NM_000094.3(COL7A1):c.6752G>A (p.Gly2251Glu)	COL7A1	3	48572941	Epidermolysis bullosa pruriginosa, autosomal dominant;Nail disorder, nonsyndromic congenital, 8;Transient bullous dermolysis of the newborn
NM_000094.3(COL7A1):c.4888C>T (p.Arg1630Ter)	COL7A1	3	48581271	Epidermolysis bullosa pruriginosa, autosomal recessive;not provided
NM_000424.3(KRT5):c.74C>T (p.Pro25Leu)	KRT5	12	52520223	Epidermolysis bullosa simplex with mottled pigmentation;not provided
NM_201384.2(PLEC):c.46C>T (p.Arg16Ter)	PLEC	8	143939416	Epidermolysis bullosa simplex with nail dystrophy
NM_000445.4(PLEC):c.913C>T (p.Gln305Ter)	PLEC	8	143934923	Epidermolysis bullosa simplex with pyloric atresia
NM_201378.3(PLEC):c.1221G>A (p.Ser407=)	PLEC	8	143933998	Epidermolysis bullosa simplex with pyloric atresia
NM_000445.4(PLEC):c.9085C>T (p.Arg3029Ter)	PLEC	8	143920817	Epidermolysis bullosa simplex with pyloric atresia
NM_000424.3(KRT5):c.1252G>A (p.Glu418Lys)	KRT5	12	52516824	Epidermolysis bullosa simplex, autosomal recessive;not provided
NM_000526.4(KRT14):c.357G>A (p.Met119Ile)	KRT14	17	41586478	Epidermolysis bullosa simplex, Cockayne-Touraine type;Epidermolysis bullosa simplex, autosomal recessive;not provided
NM_000424.3(KRT5):c.508G>A (p.Glu170Lys)	KRT5	12	52519789	Epidermolysis bullosa simplex, Cockayne-Touraine type;Epidermolysis bullosa simplex, autosomal recessive;not provided
NM_017644.3(KLHL24):c.3G>A (p.Met1Ile)	KLHL24	3	183650359	EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED, WITH SCARRING AND HAIR LOSS;Epidermolysis bullosa simplex, Koebner type
NM_201380.3(PLEC):c.6328C>T (p.Arg2110Trp)	PLEC	8	143924012	Epidermolysis bullosa simplex, Ogna type
NM_000494.3(COL17A1):c.3908G>A (p.Arg1303Gln)	COL17A1	10	104034193	Epidermolysis bullosa, junctional, localisata variant

NM_004415.3(DSP):c.5800C>T (p.Arg1934Ter)	DSP	6	7583062	Epidermolysis bullosa, lethal acantholytic
NM_000094.3(COL7A1):c.5096C>T (p.Pro1699Leu)	COL7A1	3	48580301	Epidermolysis bullosa, pretibial, autosomal recessive
NM_000226.3(KRT9):c.488G>A (p.Arg163Gln)	KRT9	17	41571505	Epidermolytic palmoplantar keratoderma;not provided
NM_000226.3(KRT9):c.511G>A (p.Val171Met)	KRT9	17	41571482	Epidermolytic palmoplantar keratoderma;not provided
NM_000226.3(KRT9):c.487C>T (p.Arg163Trp)	KRT9	17	41571506	Epidermolytic palmoplantar keratoderma;Palmoplantar keratoderma, epidermolytic, with knuckle pads;not provided
NM_018100.3(EFHC1):c.776G>A (p.Cys259Tyr)	EFHC1	6	52454147	Epilepsy juvenile absence
NM_000816.3(GABRG2):c.245G>A (p.Arg82Gln)	GABRG2	5	162093965	Epilepsy, childhood absence 2;Familial febrile seizures 8;not provided
NM_021912.4(GABRB3):c.94G>A (p.Gly32Arg)	GABRB3	15	26772759	Epilepsy, childhood absence 5
NM_021098.2(CACNA1H):c.844G>A (p.Glu282Lys)	CACNA1H	16	1200296	Epilepsy, childhood absence 6
NM_021098.2(CACNA1H):c.2491G>A (p.Val831Met)	CACNA1H	16	1205153	Epilepsy, childhood absence 6
NM_007198.3(PLPBP):c.207+1G>A	PLPBP	8	37765634	EPILEPSY, EARLY-ONSET, VITAMIN B6-DEPENDENT
NM_007198.3(PLPBP):c.260C>T (p.Pro87Leu)	PLPBP	8	37766296	EPILEPSY, EARLY-ONSET, VITAMIN B6-DEPENDENT
NM_007198.3(PLPBP):c.722G>A (p.Arg241Gln)	PLPBP	8	37777998	EPILEPSY, EARLY-ONSET, VITAMIN B6-DEPENDENT
NM_001242896.1(DEPDC5):c.1663C>T (p.Arg555Ter)	DEPDC5	22	31815209	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.4107G>A (p.Trp1369Ter)	DEPDC5	22	31893655	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.715C>T (p.Arg239Ter)	DEPDC5	22	31792765	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.982C>T (p.Arg328Ter)	DEPDC5	22	31802739	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.3259C>T (p.Arg1087Ter)	DEPDC5	22	31857548	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.1459C>T (p.Arg487Ter)	DEPDC5	22	31815005	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.2527C>T (p.Arg843Ter)	DEPDC5	22	31843106	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.4567C>T (p.Gln1523Ter)	DEPDC5	22	31906252	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.418C>T (p.Gln140Ter)	DEPDC5	22	31778103	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.193+1G>A	DEPDC5	22	31760703	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.279+1G>A	DEPDC5	22	31765061	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.435G>A (p.Trp145Ter)	DEPDC5	22	31778120	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.484-1G>A	DEPDC5	22	31783906	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.526C>T (p.Gln176Ter)	DEPDC5	22	31783949	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.624+1G>A	DEPDC5	22	31784876	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.727C>T (p.Arg243Ter)	DEPDC5	22	31792777	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.730C>T (p.Gln244Ter)	DEPDC5	22	31792780	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.856C>T (p.Arg286Ter)	DEPDC5	22	31797688	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.1114C>T (p.Gln372Ter)	DEPDC5	22	31804194	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.1264C>T (p.Arg422Ter)	DEPDC5	22	31806168	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.1393C>T (p.Gln465Ter)	DEPDC5	22	31810589	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.1555C>T (p.Gln519Ter)	DEPDC5	22	31815101	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.1759C>T (p.Arg587Ter)	DEPDC5	22	31819114	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.1909C>T (p.Arg637Ter)	DEPDC5	22	31821540	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.2620C>T (p.Arg874Ter)	DEPDC5	22	31843199	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.3046C>T (p.Gln1016Ter)	DEPDC5	22	31846858	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.3696+5G>A	DEPDC5	22	31874410	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.3802C>T (p.Arg1268Ter)	DEPDC5	22	31876262	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.3994C>T (p.Arg1332Ter)	DEPDC5	22	31879713	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.4397G>A (p.Trp1466Ter)	DEPDC5	22	31901763	Epilepsy, familial focal, with variable foci
NM_001242896.1(DEPDC5):c.4606C>T (p.Gln1536Ter)	DEPDC5	22	31906291	Epilepsy, familial focal, with variable foci

NM_001242896.1(DEPDC5):c.434G>A (p.Trp145Ter)	DEPDC5	22	31778119	Epilepsy, familial focal, with variable foci	
NM_006545.4(NPRL2):c.100C>T (p.Arg34Ter)	NPRL2	3	50350001	EPILEPSY, FAMILIAL FOCAL, WITH VARIABLE FOCI 2	
NM_006545.4(NPRL2):c.883C>T (p.Arg295Ter)	NPRL2	3	50348173	EPILEPSY, FAMILIAL FOCAL, WITH VARIABLE FOCI 2	
NM_001077350.2(NPRL3):c.1270C>T (p.Arg424Ter)	NPRL3	16	89794	EPILEPSY, FAMILIAL FOCAL, WITH VARIABLE FOCI 3	
NM_005045.3(RELN):c.9526G>A (p.Glu3176Lys)	RELN	7	103490747	Epilepsy, familial temporal lobe, 7	
NM_005045.3(RELN):c.2531C>T (p.Pro844Leu)	RELN	7	103630111	Epilepsy, familial temporal lobe, 7	
NM_005045.3(RELN):c.2015C>T (p.Pro672Leu)	RELN	7	103640597	Epilepsy, familial temporal lobe, 7	
NM_000833.4(GRIN2A):c.652C>T (p.Gln218Ter)	GRIN2A	16	9938314	Epilepsy, focal, with speech disorder and with or without mental retardation	
NM_000833.4(GRIN2A):c.1007+1G>A	GRIN2A	16	9937958	Epilepsy, focal, with speech disorder and with or without mental retardation	
NM_000833.4(GRIN2A):c.1592C>T (p.Thr531Met)	GRIN2A	16	9840706	Epilepsy, focal, with speech disorder and with or without mental retardation	
NM_000833.4(GRIN2A):c.3813G>A (p.Trp1271Ter)	GRIN2A	16	9763731	Epilepsy, focal, with speech disorder and with or without mental retardation	
NM_000833.4(GRIN2A):c.1553G>A (p.Arg518His)	GRIN2A	16	9840745	Epilepsy, focal, with speech disorder and with or without mental retardation;not provided	
NM_000833.4(GRIN2A):c.2041C>T (p.Arg681Ter)	GRIN2A	16	9822391	Epilepsy, focal, with speech disorder and with or without mental retardation;not provided	
NM_000833.4(GRIN2A):c.2146G>A (p.Ala716Thr)	GRIN2A	16	9822286	Epilepsy, focal, with speech disorder and with or without mental retardation;not provided	
NM_145207.2(SPATA5):c.298G>A (p.Ala100Thr)	SPATA5	4	122929049	Epilepsy, hearing loss, and mental retardation syndrome	
NM_145207.2(SPATA5):c.1343C>T (p.Ser448Leu)	SPATA5	4	122938134	Epilepsy, hearing loss, and mental retardation syndrome	
NM_145207.2(SPATA5):c.2531C>T (p.Ala844Val)	SPATA5	4	123313913	Epilepsy, hearing loss, and mental retardation syndrome	
NM_145207.2(SPATA5):c.556C>T (p.Arg186Ter)	SPATA5	4	122934147	Epilepsy, hearing loss, and mental retardation syndrome	
NM_000815.4(GABRD):c.659G>A (p.Arg220His)	GABRD	1	2028260	Epilepsy, idiopathic generalized 10;Epilepsy, juvenile myoclonic 7;Generalized epilepsy with febrile seizures plus type 5	
NM_021098.2(CACNA1H):c.2626G>A (p.Ala876Thr)	CACNA1H	16	1206126	Epilepsy, idiopathic generalized 6	
NM_000388.3(CASR):c.2693G>A (p.Arg898Gln)	CASR	3	122284647	Epilepsy, idiopathic generalized 8	
NM_006516.2(SLC2A1):c.997C>T (p.Arg333Trp)	SLC2A1	1	42929009	Epilepsy, idiopathic generalized, susceptibility to, 12;GLUT1 deficiency syndrome 1	
NM_006516.2(SLC2A1):c.940G>A (p.Gly314Ser)	SLC2A1	1	42929242	Epilepsy, idiopathic generalized, susceptibility to, 12;GLUT1 deficiency syndrome 1;GLUT1 deficiency syndrome 1, autosomal recessive;GLUT1 deficiency syndrome 2;not provided	
NM_006516.2(SLC2A1):c.377G>A (p.Arg126His)	SLC2A1	1	42930765	Epilepsy, idiopathic generalized, susceptibility to, 12;GLUT1 deficiency syndrome 1;not provided	
NM_006516.2(SLC2A1):c.694C>T (p.Arg232Cys)	SLC2A1	1	42929766	Epilepsy, idiopathic generalized, susceptibility to, 12;not provided	
NM_001134771.1(SLC12A5):c.2924G>A (p.Arg975His)	SLC12A5	20	46056217	Epilepsy, idiopathic generalized, susceptibility to, 14	
NM_001134771.1(SLC12A5):c.3214C>T (p.Arg1072Cys)	SLC12A5	20	46057189	Epilepsy, idiopathic generalized, susceptibility to, 14	
NM_000806.5(GABRA1):c.655G>A (p.Asp219Asn)	GABRA1	5	161882653	Epilepsy, juvenile myoclonic 5	
NM_000726.4(CACNB4):c.1444C>T (p.Arg482Ter)	CACNB4	2	151839238	Epilepsy, juvenile myoclonic 6	
NM_005097.3(LGI1):c.1418C>T (p.Ser473Leu)	LGI1	10	93797547	Epilepsy, lateral temporal lobe, autosomal dominant	
NM_005097.3(LGI1):c.1420C>T (p.Arg474Ter)	LGI1	10	93797549	Epilepsy, lateral temporal lobe, autosomal dominant	
NM_005097.3(LGI1):c.688C>T (p.Gln230Ter)	LGI1	10	93793200	Epilepsy, lateral temporal lobe, autosomal dominant	
NM_005097.3(LGI1):c.1128G>A (p.Trp376Ter)	LGI1	10	93797257	Epilepsy, lateral temporal lobe, autosomal dominant	
NM_005097.3(LGI1):c.406C>T (p.Arg136Trp)	LGI1	10	93777592	Epilepsy, lateral temporal lobe, autosomal dominant;not provided	
NM_020822.2(KCNT1):c.2782C>T (p.Arg928Cys)	KCNT1	9	135779411	Epilepsy, nocturnal frontal lobe, 5	
NM_020822.2(KCNT1):c.2688G>A (p.Met896Ile)	KCNT1	9	135778781	Epilepsy, nocturnal frontal lobe, 5	
NM_172218.2(SPAG1):c.2014C>T (p.Gln672Ter)	SPAG1	8	100233436	Epilepsy, nocturnal frontal lobe, 5;Kartagener syndrome	
NM_020822.2(KCNT1):c.1193G>A (p.Arg398Gln)	KCNT1	9	135765188	Epilepsy, nocturnal frontal lobe, 5;not provided	
NM_000744.6(CHRNA4):c.878C>T (p.Thr293Ile)	CHRNA4	20	63350533	Epilepsy, nocturnal frontal lobe, type 1	
NM_000744.6(CHRNA4):c.839C>T (p.Ser280Phe)	CHRNA4	20	63350572	Epilepsy, nocturnal frontal lobe, type 1;not provided	
NM_000744.6(CHRNA4):c.851C>T (p.Ser2841eu)	CHRNA4	20	63350560	Epilepsy, nocturnal frontal lobe, type 1;not provided	
NM_000748.2(CHRNB2):c.859G>A (p.Val287Met)	CHRNB2	1	154571682	Epilepsy, nocturnal frontal lobe, type 3	
NM_198586.2(NHLRC1):c.793C>T (p.Arg265Ter)	NHLRC1	6	18121814	Epilepsy, progressive myoclonic 2b	

NM_153033.4(KCTD7):c.295C>T (p.Arg99Ter)	KCTD7	7	66633425	Epilepsy, progressive myoclonic 3
NM_153033.4(KCTD7):c.280C>T (p.Arg94Trp)	KCTD7	7	66633410	Epilepsy, progressive myoclonic 3
NM_005506.3(SCARB2):c.862C>T (p.Gln288Ter)	SCARB2	4	76174276	Epilepsy, progressive myoclonic 4, with or without renal failure
NM_005506.3(SCARB2):c.533G>A (p.Trp178Ter)	SCARB2	4	76179596	Epilepsy, progressive myoclonic 4, with or without renal failure
NM_005506.3(SCARB2):c.1270C>T (p.Arg424Ter)	SCARB2	4	76163353	Epilepsy, progressive myoclonic 4, with or without renal failure
NM_005506.3(SCARB2):c.704+5G>A	SCARB2	4	76176432	Epilepsy, progressive myoclonic 4, with or without renal failure
NM_005506.3(SCARB2):c.704+1G>A	SCARB2	4	76176436	Epilepsy, progressive myoclonic 4, with or without renal failure
NM_005506.3(SCARB2):c.361C>T (p.Arg121Ter)	SCARB2	4	76181016	Epilepsy, progressive myoclonic 4, with or without renal failure;not provided
NM_004287.4(GOSR2):c.336+1G>A	GOSR2	17	46932200	Epilepsy, progressive myoclonic 6;not provided
NM_001112741.1(KCNC1):c.959G>A (p.Arg320His)	KCNC1	11	17772053	Epilepsy, progressive myoclonic 7
NM_153033.4(KCTD7):c.550C>T (p.Arg184Cys)	KCTD7	7	66638912	Epilepsy, progressive myoclonic, 3, with intracellular inclusions
NM_032737.3(LMNB2):c.469C>T (p.His157Tyr)	LMNB2	19	2438464	Epilepsy, progressive myoclonic, 9
NM_006950.3(SYN1):c.1067G>A (p.Trp356Ter)	SYN1	X	47576222	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
NM_006950.3(SYN1):c.1663C>T (p.Gln555Ter)	SYN1	X	47574321	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
NM_006950.3(SYN1):c.1648G>A (p.Ala550Thr)	SYN1	X	47574336	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
NM_006950.3(SYN1):c.377G>A (p.Trp126Ter)	SYN1	X	47619352	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
NM_003165.3(STXBP1):c.1565G>A (p.Trp522Ter)	STXBP1	9	127682423	Epileptic encephalopathy
NM_001271.3(CHD2):c.361C>T (p.Arg121Ter)	CHD2	15	92927310	Epileptic encephalopathy, childhood-onset
NM_001271.3(CHD2):c.4971G>A (p.Trp1657Ter)	CHD2	15	93020076	Epileptic encephalopathy, childhood-onset
NM_001271.3(CHD2):c.1502+1G>A	CHD2	15	92949077	Epileptic encephalopathy, childhood-onset
NM_001271.3(CHD2):c.1942C>T (p.Pro648Ser)	CHD2	15	92956591	Epileptic encephalopathy, childhood-onset
NM_001271.3(CHD2):c.2725C>T (p.Gln909Ter)	CHD2	15	92978381	Epileptic encephalopathy, childhood-onset
NM_001271.3(CHD2):c.4921C>T (p.Gln1641Ter)	CHD2	15	93020026	Epileptic encephalopathy, childhood-onset
NM_001271.3(CHD2):c.1396C>T (p.Arg466Ter)	CHD2	15	92948970	Epileptic encephalopathy, childhood-onset;not provided
NM_001271.3(CHD2):c.4909C>T (p.Arg1637Ter)	CHD2	15	93020014	Epileptic encephalopathy, childhood-onset;not provided
NM_139058.2(ARX):c.1058C>T (p.Pro353Leu)	ARX	X	25012937	Epileptic encephalopathy, early infantile, 1
NM_016373.3(WWOX):c.790C>T (p.Arg264Ter)	WWOX	16	78425054	Epileptic encephalopathy, early infantile, 1;Spinocerebellar ataxia, autosomal recessive 12;not provided
NM_000806.5(GABRA1):c.751G>A (p.Gly251Ser)	GABRA1	5	161890945	Epileptic encephalopathy, early infantile, 19
NM_000806.5(GABRA1):c.902G>A (p.Arg301Lys)	GABRA1	5	161895711	Epileptic encephalopathy, early infantile, 19
NM_000806.5(GABRA1):c.641G>A (p.Arg214His)	GABRA1	5	161882639	Epileptic encephalopathy, early infantile, 19;not provided
NM_033407.3(DOCK7):c.3616C>T (p.Arg1206Ter)	DOCK7	1	62529349	Epileptic encephalopathy, early infantile, 23
NM_021072.3(HCN1):c.299C>T (p.Ser100Phe)	HCN1	5	45695795	Epileptic encephalopathy, early infantile, 24
NM_021072.3(HCN1):c.835C>T (p.His279Tyr)	HCN1	5	45645199	Epileptic encephalopathy, early infantile, 24
NM_177550.4(SLC13A5):c.655G>A (p.Gly219Arg)	SLC13A5	17	6703031	Epileptic encephalopathy, early infantile, 25
NM_177550.4(SLC13A5):c.680C>T (p.Thr227Met)	SLC13A5	17	6703006	Epileptic encephalopathy, early infantile, 25
NM_177550.4(SLC13A5):c.1280C>T (p.Ser427Leu)	SLC13A5	17	6690936	Epileptic encephalopathy, early infantile, 25
NM_177550.4(SLC13A5):c.1022G>A (p.Trp341Ter)	SLC13A5	17	6695759	Epileptic encephalopathy, early infantile, 25
NM_177550.4(SLC13A5):c.425C>T (p.Thr142Met)	SLC13A5	17	6704000	Epileptic encephalopathy, early infantile, 25
NM_177550.4(SLC13A5):c.997C>T (p.Arg333Ter)	SLC13A5	17	6695784	Epileptic encephalopathy, early infantile, 25;not provided
NM_004975.3(KCNB1):c.1135G>A (p.Gly379Arg)	KCNB1	20	49374425	Epileptic encephalopathy, early infantile, 26
NM_004975.3(KCNB1):c.1121C>T (p.Thr374Ile)	KCNB1	20	49374439	Epileptic encephalopathy, early infantile, 26;not provided
NM_000834.3(GRIN2B):c.1619G>A (p.Arg540His)	GRIN2B	12	13615149	Epileptic encephalopathy, early infantile, 27
NM_016373.3(WWOX):c.160C>T (p.Arg54Ter)	WWOX	16	78108475	Epileptic encephalopathy, early infantile, 28
NM_016373.3(WWOX):c.1005G>A (p.Trp335Ter)	WWOX	16	78432701	Epileptic encephalopathy, early infantile, 28

NM_173354.4(SIK1):c.1840C>T (p.Gln614Ter)	SIK1	21	43417679	Epileptic encephalopathy, early infantile, 30	
NM_173354.4(SIK1):c.1897C>T (p.Gln633Ter)	SIK1	21	43417622	Epileptic encephalopathy, early infantile, 30	
NM_173354.4(SIK1):c.1906G>A (p.Gly636Ser)	SIK1	21	43417613	Epileptic encephalopathy, early infantile, 30	
NM_001005336.2(DNM1):c.139G>A (p.Val47Met)	DNM1	9	128203609	Epileptic encephalopathy, early infantile, 31	
NM_004974.3(KCNA2):c.1214C>T (p.Pro405Leu)	KCNA2	1	110603569	Epileptic encephalopathy, early infantile, 32;not provided	
NM_004974.3(KCNA2):c.890G>A (p.Arg297Gln)	KCNA2	1	110603893	Epileptic encephalopathy, early infantile, 32;not provided	
NM_001958.3(EEF1A2):c.364G>A (p.Glu122Lys)	EEF1A2	20	63495062	Epileptic encephalopathy, early infantile, 33;Mental retardation, autosomal dominant 38	
NM_001958.3(EEF1A2):c.208G>A (p.Gly70Ser)	EEF1A2	20	63495972	Epileptic encephalopathy, early infantile, 33;not provided	
NM_001324236.1(ITPA):c.115G>A (p.Gly39Arg)	ITPA	20	3221881	Epileptic encephalopathy, early infantile, 35	
NM_001324236.1(ITPA):c.195C>T (p.Ile65=)	ITPA	20	3223409	Epileptic encephalopathy, early infantile, 35	
NM_001127221.1(CACNA1A):c.2137G>A (p.Ala713Thr)	CACNA1A	19	13303584	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 42;not provided	
NM_021912.4(GABRB3):c.358G>A (p.Asp120Asn)	GABRB3	15	26621417	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 43	
NM_021912.4(GABRB3):c.913G>A (p.Ala305Thr)	GABRB3	15	26561099	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 43	
NM_024818.4(UBA5):c.1111G>A (p.Ala371Thr)	UBA5	3	132675903	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 44	
NM_024818.4(UBA5):c.164G>A (p.Arg55His)	UBA5	3	132665825	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 44	
NM_024818.4(UBA5):c.181C>T (p.Arg61Ter)	UBA5	3	132665842	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 44	
NM_024818.4(UBA5):c.562C>T (p.Arg188Ter)	UBA5	3	132671032	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 44	
NM_024818.4(UBA5):c.904C>T (p.Gln302Ter)	UBA5	3	132675339	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 44	
NM_024818.4(UBA5):c.778G>A (p.Val260Met)	UBA5	3	132672143	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 44	
NM_024818.4(UBA5):c.503G>A (p.Gly168Glu)	UBA5	3	132670973	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 44	
NM_000812.3(GABRB1):c.860C>T (p.Thr287Ile)	GABRB1	4	47406706	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 45	
NM_000836.2(GRIN2D):c.1999G>A (p.Val667Ile)	GRIN2D	19	48419722	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 46	
NM_021032.4(FGF12):c.341G>A (p.Arg114His)	FGF12	3	192335434	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 47;not provided	
NM_004644.4(AP3B2):c.1182G>A (p.Lys394=)	AP3B2	15	82679729	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 48	
NM_004644.4(AP3B2):c.199C>T (p.Arg67Ter)	AP3B2	15	82689223	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 48	
NM_005918.3(MDH2):c.620C>T (p.Pro207Leu)	MDH2	7	76063579	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 51	
NM_005918.3(MDH2):c.398C>T (p.Pro133Leu)	MDH2	7	76058047	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 51;Infantile encephalopathy	
NM_005918.3(MDH2):c.109G>A (p.Gly37Arg)	MDH2	7	76054872	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 51;Infantile encephalopathy	
NM_203446.2(SYNJ1):c.2528G>A (p.Trp843Ter)	SYNJ1	21	32657766	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 53	
NM_031844.2(HNRNPU):c.511C>T (p.Gln171Ter)	HNRNPU	1	244863797	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 54	
NM_021007.2(SCN2A):c.3947C>T (p.Ala1316Val)	SCN2A	2	165373322	Epileptic encephalopathy;not provided	
NM_003165.3(STXBP1):c.1216C>T (p.Arg406Cys)	STXBP1	9	127675909	Epileptic encephalopathy;not provided	
NM_003995.3(NPR2):c.2647G>A (p.Val883Met)	NPR2	9	35807333	Epiphyseal chondrodysplasia, miura type	
NM_003995.3(NPR2):c.1963C>T (p.Arg655Cys)	NPR2	9	35805586	Epiphyseal chondrodysplasia, miura type	
NM_001853.3(COL9A3):c.148-1G>A	COL9A3	20	62818517	Epiphyseal dysplasia, multiple, 3, with myopathy	
NM_033150.2(COL2A1):c.2503C>T (p.Arg835Cys)	COL2A1	12	47979534	Epiphyseal dysplasia, multiple, with myopia and conductive deafness;Myopia	
NM_000217.2(KCNA1):c.1210G>A (p.Val404Ile)	KCNA1	12	4912588	Episodic ataxia type 1	
NM_000217.2(KCNA1):c.1249C>T (p.Arg417Ter)	KCNA1	12	4912627	Episodic ataxia type 1	
NM_000217.2(KCNA1):c.677C>T (p.Thr226Met)	KCNA1	12	4912055	Episodic ataxia type 1	
NM_001127221.1(CACNA1A):c.3992+1G>A	CACNA1A	19	13275849	Episodic ataxia type 2	
NM_001127221.1(CACNA1A):c.4982G>A (p.Arg1661His)	CACNA1A	19	13235702	Episodic ataxia type 2	
NM_001127221.1(CACNA1A):c.5266G>A (p.Glu1756Lys)	CACNA1A	19	13231847	Episodic ataxia type 2	
NM_001127221.1(CACNA1A):c.3832C>T (p.Arg1278Ter)	CACNA1A	19	13277122	Episodic ataxia type 2	
NM_001127221.1(CACNA1A):c.4636C>T (p.Arg1546Ter)	CACNA1A	19	13255217	Episodic ataxia type 2	
NM_001127221.1(CACNA1A):c.860G>A (p.Cys287Tyr)	CACNA1A	19	13359724	Episodic ataxia type 2	

NM_001127221.1(CACNA1A):c.3825+1G>A	CACNA1A	19	13283266	Episodic ataxia type 2
NM_001127221.1(CACNA1A):c.877G>A (p.Gly293Arg)	CACNA1A	19	13359707	Episodic ataxia type 2;Spinocerebellar ataxia 6
NM_001127221.1(CACNA1A):c.5455C>T (p.Arg1819Ter)	CACNA1A	19	13230158	Episodic ataxia, type 2, and epilepsy
NM_004172.4(SLC1A3):c.1496G>A (p.Arg499Gln)	SLC1A3	5	36686136	Episodic ataxia, type 6
NM_015488.4(PNKD):c.26C>T (p.Ala9Val)	PNKD	2	218270561	Episodic hemiplegia;Paroxysmal choreoathetosis;Paroxysmal dyskinesia;Paroxysmal dystonia
NM_006514.3(SCN10A):c.3910G>A (p.Ala1304Thr)	SCN10A	3	38712340	Episodic pain syndrome, familial, 2
NM_001349253.1(SCN11A):c.673C>T (p.Arg225Cys)	SCN11A	3	38925454	Episodic pain syndrome, familial, 3
NM_000020.2(ACVRL1):c.1451G>A (p.Arg484Gln)	ACVRL1	12	51920832	Epistaxis;Pulmonary arterial hypertension;not provided
NM_000494.3(COL17A1):c.2816C>T (p.Thr939Ile)	COL17A1	10	104039613	Epithelial recurrent erosion dystrophy
NM_000494.3(COL17A1):c.3156C>T (p.Gly1052=)	COL17A1	10	104037688	Epithelial recurrent erosion dystrophy
NM_002473.5(MYH9):c.2105G>A (p.Arg702His)	MYH9	22	36305984	Epstein syndrome;Fechtner syndrome;MYH9 related disorders
NM_002473.5(MYH9):c.2104C>T (p.Arg702Cys)	MYH9	22	36305985	Epstein syndrome;Fechtner syndrome;MYH9 related disorders;May-Hegglin anomaly;Sebastian syndrome
NM_000551.3(VHL):c.499C>T (p.Arg167Trp)	VHL	3	10149822	Erythrocytosis, familial, 2;Hereditary cancer-predisposing syndrome;Pheochromocytoma;Renal cell carcinoma;Von Hippel-Lindau syndrome;not provided
NM_000551.3(VHL):c.500G>A (p.Arg167Gln)	VHL	3	10149823	Erythrocytosis, familial, 2;Hereditary cancer-predisposing syndrome;Von Hippel-Lindau syndrome;not provided
NM_000551.3(VHL):c.481C>T (p.Arg161Ter)	VHL	3	10149804	Erythrocytosis, familial, 2;Renal cell carcinoma;Von Hippel-Lindau syndrome;not provided
NM_000551.3(VHL):c.337C>T (p.Arg113Ter)	VHL	3	10142184	Erythrocytosis, familial, 2;Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.482G>A (p.Arg161Gln)	VHL	3	10149805	Erythrocytosis, familial, 2;Von Hippel-Lindau syndrome;not provided
NM_022051.2(EGLN1):c.1112G>A (p.Arg371His)	EGLN1	1	231370598	Erythrocytosis, familial, 3
NM_001430.4(EPAS1):c.1609G>A (p.Gly537Arg)	EPAS1	2	46380281	Erythrocytosis, familial, 4
NM_001942.3(DSG1):c.49-1G>A	DSG1	18	31326580	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper-ige
NM_000421.3(KRT10):c.1373+1G>A	KRT10	17	40819516	Erythroderma, ichthyosiform, congenital reticular
NM_153212.2(GJB4):c.35G>A (p.Gly12Asp)	GJB4	1	34761289	Erythrokeratodermia variabilis
NM_153212.2(GJB4):c.65G>A (p.Arg22His)	GJB4	1	34761319	Erythrokeratodermia variabilis
NM_024009.2(GJB3):c.35G>A (p.Gly12Asp)	GJB3	1	34784797	Erythrokeratodermia variabilis
NM_000165.4(GJA1):c.131C>T (p.Ala44Val)	GJA1	6	121446978	Erythrokeratodermia variabilis
NM_000140.3(FECH):c.1077+1G>A	FECH	18	57554259	Erythropoietic protoporphyria
NM_001012515.2(FECH):c.571G>A (p.Ala191Thr)	FECH	18	57566492	Erythropoietic protoporphyria
NM_005977.3(RNF6):c.305G>A (p.Arg102Lys)	RNF6	13	26215577	Esophageal squamous cell carcinoma, somatic
NM_005977.3(RNF6):c.724G>A (p.Ala242Thr)	RNF6	13	26215158	Esophageal squamous cell carcinoma, somatic
NM_005977.3(RNF6):c.731G>A (p.Gly244Asp)	RNF6	13	26215151	Esophageal squamous cell carcinoma, somatic
NM_005373.2(MPL):c.1514G>A (p.Ser505Asn)	MPL	1	43349308	Essential thrombocythemia;Myelofibrosis;Myeloproliferative Neoplasm;Thrombocytosis, benign familial microcytic
NM_001122742.1(ESR1):c.469C>T (p.Arg157Ter)	ESR1	6	151842613	Estrogen resistance
NM_014297.4(ETHE1):c.487C>T (p.Arg163Trp)	ETHE1	19	43511455	Ethylmalonic encephalopathy
NM_004134.6(HSPA9):c.376C>T (p.Arg126Trp)	HSPA9	5	138570994	Even-plus syndrome
m.15615G>A	MT-CYB	MT	15615	Exercise intolerance
m.14846G>A	MT-CYB	MT	14846	Exercise intolerance
m.15150G>A	MT-CYB	MT	15150	Exercise intolerance
m.7497G>A	MT-TS1	MT	7497	Exercise intolerance, muscle pain, and lactic acidemia
NM_030780.4(SLC25A32):c.425G>A (p.Trp142Ter)	SLC25A32	8	103403291	EXERCISE INTOLERANCE, RIBOFLAVIN-RESPONSIVE (1 patient)
NM_030780.4(SLC25A32):c.440G>A (p.Arg147His)	SLC25A32	8	103403276	EXERCISE INTOLERANCE, RIBOFLAVIN-RESPONSIVE (1 patient)
NM_003051.3(SLC16A1):c.-202G>A	SLC16A1	1	112956192	Exercise-induced hyperinsulinemic hypoglycemia

NM_005213.3(CSTA):c.256C>T (p.Gln86Ter)	CSTA	3	122341526	Exfoliative ichthyosis, autosomal recessive, ichthyosis bullosa of siemens-like	
NM_005213.3(CSTA):c.172C>T (p.Arg58Ter)	CSTA	3	122341442	Exfoliative ichthyosis, autosomal recessive, ichthyosis bullosa of siemens-like	
NM_001904.3(CTNNB1):c.2128C>T (p.Arg710Cys)	CTNNB1	3	41238067	Exudative vitreoretinopathy 1	
NM_012193.3(FZD4):c.1250G>A (p.Arg417Gln)	FZD4	11	86951506	Exudative vitreoretinopathy 1;Exudative vitreoretinopathy, digenic	
NM_002335.3(LRP5):c.433C>T (p.Leu145Phe)	LRP5	11	68348188	Exudative vitreoretinopathy 4, autosomal dominant;Osteoporosis with pseudoglioma	
NM_002335.3(LRP5):c.1709G>A (p.Arg570Gln)	LRP5	11	68403607	Exudative vitreoretinopathy 4, autosomal recessive	
NM_002335.3(LRP5):c.4099G>A (p.Glu1367Lys)	LRP5	11	68436987	Exudative vitreoretinopathy 4, autosomal recessive	
NM_002335.3(LRP5):c.1828G>A (p.Gly610Arg)	LRP5	11	68406550	Exudative vitreoretinopathy 4, autosomal recessive	
NM_002335.3(LRP5):c.1330C>T (p.Arg444Cys)	LRP5	11	68386630	Exudative vitreoretinopathy 4, digenic	
NM_012338.3(TSPAN12):c.146C>T (p.Thr49Met)	TSPAN12	7	120840030	Exudative vitreoretinopathy 5	
NM_012338.3(TSPAN12):c.285+1G>A	TSPAN12	7	120838776	Exudative vitreoretinopathy 5	
NM_024741.2(ZNF408):c.1363C>T (p.His455Tyr)	ZNF408	11	46705063	Exudative vitreoretinopathy 6	
NM_000169.2(GLA):c.1066C>T (p.Arg356Trp)	GLA	X	101398033	Fabry disease	
NM_000169.2(GLA):c.131G>A (p.Trp44Ter)	GLA	X	101407773	Fabry disease	
NM_000169.2(GLA):c.118C>T (p.Pro40Ser)	GLA	X	101407786	Fabry disease	
NM_000169.2(GLA):c.982G>A (p.Gly328Arg)	GLA	X	101398387	Fabry disease	
NM_000169.2(GLA):c.436C>T (p.Pro146Ser)	GLA	X	101401743	Fabry disease	
NM_000169.2(GLA):c.466G>A (p.Ala156Thr)	GLA	X	101401713	Fabry disease	
NM_000169.2(GLA):c.861G>A (p.Trp287Ter)	GLA	X	101398508	Fabry disease	
NM_000169.2(GLA):c.890C>T (p.Ser297Phe)	GLA	X	101398479	Fabry disease	
NM_000169.2(GLA):c.1020G>A (p.Trp340Ter)	GLA	X	101398079	Fabry disease	
NM_000169.2(GLA):c.1025G>A (p.Arg342Gln)	GLA	X	101398074	Fabry disease	
NM_000169.2(GLA):c.1024C>T (p.Arg342Ter)	GLA	X	101398075	Fabry disease	
NM_000169.2(GLA):c.281G>A (p.Cys94Tyr)	GLA	X	101403899	Fabry disease	
NM_000169.2(GLA):c.677G>A (p.Trp226Ter)	GLA	X	101398909	Fabry disease	
NM_000169.2(GLA):c.734G>A (p.Trp245Ter)	GLA	X	101398852	Fabry disease	
NM_000169.2(GLA):c.748C>T (p.Gln250Ter)	GLA	X	101398838	Fabry disease	
NM_000169.2(GLA):c.658C>T (p.Arg220Ter)	GLA	X	101398928	Fabry disease	
NM_000169.2(GLA):c.730G>A (p.Asp244Asn)	GLA	X	101398856	Fabry disease	
NM_000169.2(GLA):c.369+1G>A	GLA	X	101403810	Fabry disease	
NM_000169.2(GLA):c.335G>A (p.Arg112His)	GLA	X	101403845	Fabry disease	
NM_000169.2(GLA):c.485G>A (p.Trp162Ter)	GLA	X	101401694	Fabry disease	
NM_000169.2(GLA):c.661C>T (p.Gln221Ter)	GLA	X	101398925	Fabry disease	
NM_000169.2(GLA):c.916C>T (p.Gln306Ter)	GLA	X	101398453	Fabry disease	
NM_000169.2(GLA):c.1072G>A (p.Glu358Lys)	GLA	X	101398027	Fabry disease	
NM_000169.2(GLA):c.1087C>T (p.Arg363Cys)	GLA	X	101398012	Fabry disease	
NM_000169.2(GLA):c.1088G>A (p.Arg363His)	GLA	X	101398011	Fabry disease	
NM_000169.2(GLA):c.605G>A (p.Cys202Tyr)	GLA	X	101400700	Fabry disease	
NM_000169.2(GLA):c.830G>A (p.Trp277Ter)	GLA	X	101398539	Fabry disease	
NM_000169.2(GLA):c.979C>T (p.Gln327Ter)	GLA	X	101398390	Fabry disease	
NM_000169.2(GLA):c.422C>T (p.Thr141Ile)	GLA	X	101401757	Fabry disease	
NM_000169.2(GLA):c.285G>A (p.Trp95Ter)	GLA	X	101403895	Fabry disease	
NM_000169.2(GLA):c.735G>A (p.Trp245Ter)	GLA	X	101398851	Fabry disease	
NM_000169.2(GLA):c.888G>A (p.Met296Ile)	GLA	X	101398481	Fabry disease, cardiac variant	
NM_000169.2(GLA):c.639+919G>A	GLA	X	101399747	Fabry disease;Fabry disease, cardiac variant	

NM_000169.2(GLA):c.680G>A (p.Arg227Gln)	GLA	X	101398906	Fabry disease;not provided
NM_000169.2(GLA):c.679C>T (p.Arg227Ter)	GLA	X	101398907	Fabry disease;not provided
NM_000169.2(GLA):c.334C>T (p.Arg112Cys)	GLA	X	101403846	Fabry disease;not provided
NM_000169.2(GLA):c.242G>A (p.Trp81Ter)	GLA	X	101403938	Fabry disease;not provided
NM_000169.2(GLA):c.901C>T (p.Arg301Ter)	GLA	X	101398468	Fabry disease;not provided
NM_000169.2(GLA):c.974G>A (p.Gly325Asp)	GLA	X	101398395	Fabry disease;not provided
NM_000169.2(GLA):c.847C>T (p.Gln283Ter)	GLA	X	101398522	Fabry disease;not provided
NM_000169.2(GLA):c.469C>T (p.Gln157Ter)	GLA	X	101401710	Fabry disease;not provided
NM_000169.2(GLA):c.1118G>A (p.Gly373Asp)	GLA	X	101397981	Fabry disease;not provided
NM_004318.3(ASPH):c.2203C>T (p.Arg735Trp)	ASPH	8	61503433	Facial dysmorphism, lens dislocation, anterior segment abnormalities, and spontaneous filtering blebs
NM_015295.2(SMCHD1):c.2068C>T (p.Pro690Ser)	SMCHD1	18	2707567	Facioscapulohumeral muscular dystrophy 2
NM_015295.2(SMCHD1):c.1580C>T (p.Thr527Met)	SMCHD1	18	2700851	Facioscapulohumeral muscular dystrophy 2
NM_015295.2(SMCHD1):c.3801+1G>A	SMCHD1	18	2743929	Facioscapulohumeral muscular dystrophy 2
NM_015295.2(SMCHD1):c.1843-1G>A	SMCHD1	18	2705693	Facioscapulohumeral muscular dystrophy 2
NM_000186.3(CFH):c.2876G>A (p.Cys959Tyr)	CFH	1	196740712	Factor H deficiency
NM_001171507.2(MCFD2):c.149+5G>A	MCFD2	2	46909018	Factor v and factor viii, combined deficiency of, 2
NM_001171507.2(MCFD2):c.309+1G>A	MCFD2	2	46907809	Factor v and factor viii, combined deficiency of, 2
NM_000130.4(F5):c.2401C>T (p.Gln801Ter)	F5	1	169542689	Factor V deficiency
NM_000130.4(F5):c.3481C>T (p.Arg1161Ter)	F5	1	169541609	Factor V deficiency
NM_000130.4(F5):c.6304C>T (p.Arg2102Cys)	F5	1	169518453	Factor V deficiency
NM_000131.4(F7):c.1061C>T (p.Ala354Val)	F7	13	113118668	Factor VII deficiency
NM_000131.4(F7):c.628C>T (p.Gln210Ter)	F7	13	113116822	Factor VII deficiency
NM_000504.3(F10):c.1096C>T (p.Arg366Cys)	F10	13	113149146	Factor X deficiency
NM_000504.3(F10):c.424G>A (p.Glu142Lys)	F10	13	113140972	Factor X deficiency
NM_000129.3(F13A1):c.2045G>A (p.Arg682His)	F13A1	6	6151813	Factor xiii, a subunit, deficiency of
NM_000129.3(F13A1):c.514C>T (p.Arg172Ter)	F13A1	6	6266615	Factor xiii, a subunit, deficiency of
NM_000129.3(F13A1):c.1504G>A (p.Gly502Arg)	F13A1	6	6174823	Factor xiii, a subunit, deficiency of
NM_000129.3(F13A1):c.1687G>A (p.Gly563Arg)	F13A1	6	6174640	Factor xiii, a subunit, deficiency of
NM_000129.3(F13A1):c.980G>A (p.Arg327Gln)	F13A1	6	6222165	Factor xiii, a subunit, deficiency of
NM_000129.3(F13A1):c.2110C>T (p.Arg704Trp)	F13A1	6	6145708	Factor xiii, a subunit, deficiency of
NM_000129.3(F13A1):c.1984C>T (p.Arg662Ter)	F13A1	6	6151874	Factor xiii, a subunit, deficiency of
NM_001079812.2(DIAPH1):c.2305C>T (p.Gln769Ter)	DIAPH1	5	141573518	Failure to thrive;Global developmental delay;Microcephaly;Seizure disorder;Seizures, cortical blindness, and microcephaly syndrome
NM_015331.2(NCSTN):c.349C>T (p.Arg117Ter)	NCSTN	1	160349583	Familial acne inversa 1
NM_000038.5(APC):c.1690C>T (p.Arg564Ter)	APC	5	112828919	Familial adenomatous polyposis 1
NM_000038.5(APC):c.470G>A (p.Trp157Ter)	APC	5	112775676	Familial adenomatous polyposis 1
NM_000038.5(APC):c.423-1G>A	APC	5	112775628	Familial adenomatous polyposis 1
NM_000038.5(APC):c.1262G>A (p.Trp421Ter)	APC	5	112819294	Familial adenomatous polyposis 1
NM_000038.5(APC):c.1072C>T (p.Gln358Ter)	APC	5	112819104	Familial adenomatous polyposis 1
NM_001127511.2(APC):c.-190G>A	APC	5	112707528	Familial adenomatous polyposis 1
NM_000038.5(APC):c.2096G>A (p.Trp699Ter)	APC	5	112837690	Familial adenomatous polyposis 1
NM_000038.5(APC):c.471G>A (p.Trp157Ter)	APC	5	112775677	Familial adenomatous polyposis 1
NM_000038.5(APC):c.2097G>A (p.Trp699Ter)	APC	5	112837691	Familial adenomatous polyposis 1
NM_000038.5(APC):c.2635C>T (p.Gln879Ter)	APC	5	112838229	Familial adenomatous polyposis 1

NM_000038.5(APC):c.1495C>T (p.Arg499Ter)	APC	5	112827194	Familial adenomatous polyposis 1;Gardner syndrome
NM_000038.5(APC):c.2413C>T (p.Arg805Ter)	APC	5	112838007	Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.481C>T (p.Gln161Ter)	APC	5	112775687	Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.1213C>T (p.Arg405Ter)	APC	5	112819245	Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.646C>T (p.Arg216Ter)	APC	5	112792446	Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.637C>T (p.Arg213Ter)	APC	5	112780895	Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.70C>T (p.Arg24Ter)	APC	5	112754960	Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.847C>T (p.Arg283Ter)	APC	5	112815507	Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.994C>T (p.Arg332Ter)	APC	5	112819026	Familial adenomatous polyposis 1;Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.694C>T (p.Arg232Ter)	APC	5	112792494	Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.1659G>A (p.Trp553Ter)	APC	5	112828888	Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.2365C>T (p.Gln789Ter)	APC	5	112837959	Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.3982C>T (p.Gln1328Ter)	APC	5	112839576	Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.3286C>T (p.Gln1096Ter)	APC	5	112838880	Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.1987C>T (p.Gln663Ter)	APC	5	112837581	Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.645+1G>A	APC	5	112780904	Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.1658G>A (p.Trp553Ter)	APC	5	112828887	Familial adenomatous polyposis 1;not provided
NM_000038.5(APC):c.3391C>T (p.Gln1131Ter)	APC	5	112838985	Familial adenomatous polyposis 1;not provided
NM_002528.6(NTHL1):c.268C>T (p.Gln90Ter)	NTHL1	16	2046238	Familial adenomatous polyposis 3
NM_002528.6(NTHL1):c.709+1G>A	NTHL1	16	2043566	Familial adenomatous polyposis 3
NM_002439.4(MSH3):c.2319-1G>A	MSH3	5	80778719	FAMILIAL ADENOMATOUS POLYPOSIS 4
NM_001243133.1(NLRP3):c.1055C>T (p.Ala352Val)	NLRP3	1	247424504	Familial amyloid nephropathy with urticaria AND deafness;Familial cold urticaria
NM_001243133.1(NLRP3):c.778C>T (p.Arg260Trp)	NLRP3	1	247424227	Familial amyloid nephropathy with urticaria AND deafness;Familial cold urticaria;not provided
NM_001243133.1(NLRP3):c.1043C>T (p.Thr348Met)	NLRP3	1	247424492	Familial amyloid nephropathy with urticaria AND deafness;Familial cold urticaria;not provided
NM_001002841.1(MYL4):c.31G>A (p.Glu11Lys)	MYL4	17	47209453	Familial atrial fibrillation
NM_003639.4(IKBKG):c.956G>A (p.Arg319Gln)	IKBKG	X	154563602	Familial atypical mycobacteriosis, type 1, X-linked
NM_001001486.1(ATP2C1):c.910G>A (p.Ala304Thr)	ATP2C1	3	130963981	Familial benign pemphigus
NM_001001486.1(ATP2C1):c.1402C>T (p.Arg468Ter)	ATP2C1	3	130969385	Familial benign pemphigus
NM_002875.4(RAD51):c.449G>A (p.Arg150Gln)	RAD51	15	40718818	Familial cancer of breast
NM_024675.3(PALB2):c.2761C>T (p.Gln921Ter)	PALB2	16	23624082	Familial cancer of breast
NM_032043.2(BRIP1):c.2447G>A (p.Trp816Ter)	BRIP1	17	61715996	Familial cancer of breast
NM_024675.3(PALB2):c.3271C>T (p.Gln1091Ter)	PALB2	16	23607943	Familial cancer of breast
NM_024675.3(PALB2):c.3420G>A (p.Trp1140Ter)	PALB2	16	23603600	Familial cancer of breast
NM_007194.3(CHEK2):c.1072C>T (p.Gln358Ter)	CHEK2	22	28696924	Familial cancer of breast
NM_000465.3(BARD1):c.526C>T (p.Gln176Ter)	BARD1	2	214781348	Familial cancer of breast
NM_024675.3(PALB2):c.1435C>T (p.Gln479Ter)	PALB2	16	23635111	Familial cancer of breast
NM_024675.3(PALB2):c.2711G>A (p.Trp904Ter)	PALB2	16	23626273	Familial cancer of breast
NM_024675.3(PALB2):c.1048C>T (p.Gln350Ter)	PALB2	16	23635498	Familial cancer of breast
NM_032043.2(BRIP1):c.2479C>T (p.Gln827Ter)	BRIP1	17	61715964	Familial cancer of breast;Fanconi anemia, complementation group J
NM_032043.2(BRIP1):c.1495C>T (p.Gln499Ter)	BRIP1	17	61784403	Familial cancer of breast;Fanconi anemia, complementation group J
NM_032043.2(BRIP1):c.1343G>A (p.Trp448Ter)	BRIP1	17	61793727	Familial cancer of breast;Fanconi anemia, complementation group J;Hereditary cancer-predisposing syndrome
NM_032043.2(BRIP1):c.918+1G>A	BRIP1	17	61808466	Familial cancer of breast;Fanconi anemia, complementation group J;Hereditary cancer-predisposing syndrome;not provided

NM_032043.2(BRIP1):c.484C>T (p.Arg162Ter)	BRIP1	17	61849152	Familial cancer of breast;Fanconi anemia, complementation group J;Hereditary cancer-predisposing syndrome;not provided	
NM_007294.3(BRCA1):c.616C>T (p.Gln206Ter)	BRCA1	17	43095900	Familial cancer of breast;Hereditary cancer-predisposing syndrome	
NM_007194.3(CHEK2):c.151C>T (p.Gln51Ter)	CHEK2	22	28734571	Familial cancer of breast;Hereditary cancer-predisposing syndrome	
NM_000465.3(BARD1):c.298C>T (p.Gln100Ter)	BARD1	2	214792363	Familial cancer of breast;Hereditary cancer-predisposing syndrome	
NM_024675.3(PALB2):c.940C>T (p.Gln314Ter)	PALB2	16	23635606	Familial cancer of breast;Hereditary cancer-predisposing syndrome	
NM_024675.3(PALB2):c.196C>T (p.Gln66Ter)	PALB2	16	23637865	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_024675.3(PALB2):c.751C>T (p.Gln251Ter)	PALB2	16	23635795	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_000465.3(BARD1):c.1690C>T (p.Gln564Ter)	BARD1	2	214745842	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_024675.3(PALB2):c.1240C>T (p.Arg414Ter)	PALB2	16	23635306	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_032043.2(BRIP1):c.627+1G>A	BRIP1	17	61847100	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_007194.3(CHEK2):c.283C>T (p.Arg95Ter)	CHEK2	22	28734439	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_000465.3(BARD1):c.1921C>T (p.Arg641Ter)	BARD1	2	214730491	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_007194.3(CHEK2):c.279G>A (p.Trp93Ter)	CHEK2	22	28734443	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_024675.3(PALB2):c.2257C>T (p.Arg753Ter)	PALB2	16	23629897	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_000465.3(BARD1):c.1996C>T (p.Gln666Ter)	BARD1	2	214730416	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_000465.3(BARD1):c.448C>T (p.Arg150Ter)	BARD1	2	214781426	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_007194.3(CHEK2):c.409C>T (p.Arg137Ter)	CHEK2	22	28725278	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_000465.3(BARD1):c.334C>T (p.Arg112Ter)	BARD1	2	214792327	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_007194.3(CHEK2):c.85C>T (p.Gln29Ter)	CHEK2	22	28734637	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_007194.3(CHEK2):c.1232G>A (p.Trp411Ter)	CHEK2	22	28695737	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_007194.3(CHEK2):c.1486C>T (p.Gln496Ter)	CHEK2	22	28689191	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_024675.3(PALB2):c.3491G>A (p.Trp1164Ter)	PALB2	16	23603529	Familial cancer of breast;Hereditary cancer-predisposing syndrome;not provided	
NM_024675.3(PALB2):c.3256C>T (p.Arg1086Ter)	PALB2	16	23607958	Familial cancer of breast;Hereditary cancer-predisposing syndrome;Pancreatic cancer 3;not provided	
NM_024675.3(PALB2):c.2718G>A (p.Trp906Ter)	PALB2	16	23626266	Familial cancer of breast;not provided	
NM_024675.3(PALB2):c.451C>T (p.Gln151Ter)	PALB2	16	23636095	Familial cancer of breast;not provided	
NM_007194.3(CHEK2):c.58C>T (p.Gln20Ter)	CHEK2	22	28734664	Familial cancer of breast;not provided;not specified	
NM_000257.3(MYH7):c.2686G>A (p.Asp896Asn)	MYH7	14	23424143	Familial cardiomyopathy	
NM_000257.3(MYH7):c.2163-1G>A	MYH7	14	23425819	Familial cardiomyopathy	
NM_144687.3(NLRP12):c.850C>T (p.Arg284Ter)	NLRP12	19	53810809	Familial cold autoinflammatory syndrome 2;not provided	
NM_001243133.1(NLRP3):c.1316C>T (p.Ala439Val)	NLRP3	1	247424765	Familial cold urticaria;not provided	
NM_001243133.1(NLRP3):c.910G>A (p.Glu304Lys)	NLRP3	1	247424359	Familial cold urticaria;not provided	
m.8009G>A	MT-CO2	MT	8009	Familial colorectal cancer	
m.6264G>A	MT-CO1	MT	6264	Familial colorectal cancer	
m.6277G>A	MT-CO1	MT	6277	Familial colorectal cancer	
m.14985G>A	MT-CYB	MT	14985	Familial colorectal cancer	
NM_003640.4(IKBKAP):c.2741C>T (p.Pro914Leu)	IKBKAP	9	108894062	Familial dysautonomia	
NM_000121.3(EPOR):c.1317G>A (p.Trp439Ter)	EPOR	19	11378194	Familial erythrocytosis, 1	
NM_000121.3(EPOR):c.1316G>A (p.Trp439Ter)	EPOR	19	11378195	Familial erythrocytosis, 1	
NM_000266.3(NDP):c.370C>T (p.Leu124Phe)	NDP	X	43949831	Familial exudative vitreoretinopathy, X-linked	
NM_000266.3(NDP):c.361C>T (p.Arg121Trp)	NDP	X	43949840	Familial exudative vitreoretinopathy, X-linked;not provided	
NM_001127221.1(CACNA1A):c.575G>A (p.Arg192Gln)	CACNA1A	19	13371744	Familial hemiplegic migraine type 1	
NM_001127221.1(CACNA1A):c.653C>T (p.Ser218Leu)	CACNA1A	19	13365448	Familial hemiplegic migraine type 1	
NM_001127221.1(CACNA1A):c.4037G>A (p.Arg1346Gln)	CACNA1A	19	13262789	Familial hemiplegic migraine type 1	
NM_001127221.1(CACNA1A):c.4999C>T (p.Arg1667Trp)	CACNA1A	19	13235685	Familial hemiplegic migraine type 1	

NM_001127221.1(CACNA1A):c.1997C>T (p.Thr666Met)	CACNA1A	19	13303877	Familial hemiplegic migraine type 1;Migraine, sporadic hemiplegic, with progressive cerebellar ataxia
NM_001127221.1(CACNA1A):c.1748G>A (p.Arg583Gln)	CACNA1A	19	13308452	Familial hemiplegic migraine type 1;Migraine, sporadic hemiplegic;Spinocerebellar ataxia 6
NM_000702.3(ATP1A2):c.901G>A (p.Gly301Arg)	ATP1A2	1	160127704	Familial hemiplegic migraine type 2
NM_000702.3(ATP1A2):c.2066G>A (p.Arg689Gln)	ATP1A2	1	160135246	Familial hemiplegic migraine type 2
NM_000702.3(ATP1A2):c.2152G>A (p.Asp718Asn)	ATP1A2	1	160135470	Familial hemiplegic migraine type 2
NM_000702.3(ATP1A2):c.2936C>T (p.Pro979Leu)	ATP1A2	1	160139735	Familial hemiplegic migraine type 2
NM_000702.3(ATP1A2):c.1244C>T (p.Thr415Met)	ATP1A2	1	160129007	Familial hemiplegic migraine type 2
NM_000702.3(ATP1A2):c.1127C>T (p.Thr376Met)	ATP1A2	1	160128761	Familial hemiplegic migraine type 2
NM_000702.3(ATP1A2):c.571G>A (p.Val191Met)	ATP1A2	1	160124371	Familial hemiplegic migraine type 2
NM_001083116.1(PRF1):c.673C>T (p.Arg225Trp)	PRF1	10	70599048	Familial hemophagocytic lymphohistiocytosis;Hemophagocytic lymphohistiocytosis, familial, 2
NM_199242.2(UNC13D):c.1389+1G>A	UNC13D	17	75836338	Familial hemophagocytic lymphohistiocytosis;Hemophagocytic lymphohistiocytosis, familial, 3;not provided
NM_000890.3(KCNJ5):c.452G>A (p.Gly151Glu)	KCNJ5	11	128911725	Familial hyperaldosteronism type 3
NM_000890.3(KCNJ5):c.736G>A (p.Glu246Lys)	KCNJ5	11	128912009	Familial hyperaldosteronism type 3
NM_000890.3(KCNJ5):c.451G>A (p.Gly151Arg)	KCNJ5	11	128911724	Familial hyperaldosteronism type 3;not provided
NM_000527.4(LDLR):c.97C>T (p.Gln33Ter)	LDLR	19	11100252	Familial hypercholesterolemia
NM_000527.4(LDLR):c.682G>A (p.Glu228Lys)	LDLR	19	11105588	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1285G>A (p.Val429Met)	LDLR	19	11113376	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1291G>A (p.Ala431Thr)	LDLR	19	11113382	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1637G>A (p.Gly546Asp)	LDLR	19	11116144	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2439G>A (p.Trp813Ter)	LDLR	19	11129562	Familial hypercholesterolemia
NM_000527.4(LDLR):c.670G>A (p.Asp224Asn)	LDLR	19	11105576	Familial hypercholesterolemia
NM_000527.4(LDLR):c.523G>A (p.Asp175Asn)	LDLR	19	11105429	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2531G>A (p.Gly844Asp)	LDLR	19	11129654	Familial hypercholesterolemia
NM_000527.4(LDLR):c.551G>A (p.Cys184Tyr)	LDLR	19	11105457	Familial hypercholesterolemia
NM_000527.4(LDLR):c.131G>A (p.Trp44Ter)	LDLR	19	11100286	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2140+1G>A	LDLR	19	11120523	Familial hypercholesterolemia
NM_000527.4(LDLR):c.621C>T (p.Gly207=)	LDLR	19	11105527	Familial hypercholesterolemia
NM_001979.5(EPHX2):c.860G>A (p.Arg287Gln)	EPHX2	8	27516348	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1978C>T (p.Gln660Ter)	LDLR	19	11120224	Familial hypercholesterolemia
NM_000527.4(LDLR):c.224G>A (p.Cys75Tyr)	LDLR	19	11102697	Familial hypercholesterolemia
NM_000527.4(LDLR):c.251C>T (p.Pro84Leu)	LDLR	19	11102724	Familial hypercholesterolemia
NM_000527.4(LDLR):c.253C>T (p.Gln85Ter)	LDLR	19	11102726	Familial hypercholesterolemia
NM_000527.4(LDLR):c.266G>A (p.Cys89Tyr)	LDLR	19	11102739	Familial hypercholesterolemia
NM_000527.4(LDLR):c.304C>T (p.Gln102Ter)	LDLR	19	11102777	Familial hypercholesterolemia
NM_000527.4(LDLR):c.311G>A (p.Cys104Tyr)	LDLR	19	11102784	Familial hypercholesterolemia
NM_000527.4(LDLR):c.326G>A (p.Cys109Tyr)	LDLR	19	11105232	Familial hypercholesterolemia
NM_000527.4(LDLR):c.661G>A (p.Asp221Asn)	LDLR	19	11105567	Familial hypercholesterolemia
NM_000527.4(LDLR):c.796G>A (p.Asp266Asn)	LDLR	19	11106666	Familial hypercholesterolemia
NM_000527.4(LDLR):c.917C>T (p.Ser306Leu)	LDLR	19	11107491	Familial hypercholesterolemia
NM_000527.4(LDLR):c.938G>A (p.Cys313Tyr)	LDLR	19	11107512	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1033C>T (p.Gln345Ter)	LDLR	19	11110744	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1048C>T (p.Arg350Ter)	LDLR	19	11110759	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1073G>A (p.Cys358Tyr)	LDLR	19	11111526	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1187-10G>A	LDLR	19	11113268	Familial hypercholesterolemia

NM_000527.4(LDLR):c.1216C>T (p.Arg406Trp)	LDLR	19	11113307	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1448G>A (p.Trp483Ter)	LDLR	19	11113624	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1469G>A (p.Trp490Ter)	LDLR	19	11113645	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1618G>A (p.Ala540Thr)	LDLR	19	11116125	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1685G>A (p.Trp562Ter)	LDLR	19	11116192	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1705+1G>A	LDLR	19	11116213	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1731G>A (p.Trp577Ter)	LDLR	19	11116884	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1898G>A (p.Arg633His)	LDLR	19	11120144	Familial hypercholesterolemia
NM_000527.4(LDLR):c.12G>A (p.Trp4Ter)	LDLR	19	11089560	Familial hypercholesterolemia
NM_000527.4(LDLR):c.11G>A (p.Trp4Ter)	LDLR	19	11089559	Familial hypercholesterolemia
NM_000527.4(LDLR):c.103C>T (p.Gln35Ter)	LDLR	19	11100258	Familial hypercholesterolemia
NM_000527.4(LDLR):c.157C>T (p.Gln53Ter)	LDLR	19	11100312	Familial hypercholesterolemia
NM_000527.4(LDLR):c.191-1G>A	LDLR	19	11102663	Familial hypercholesterolemia
NM_000527.4(LDLR):c.260G>A (p.Trp87Ter)	LDLR	19	11102733	Familial hypercholesterolemia
NM_000527.4(LDLR):c.261G>A (p.Trp87Ter)	LDLR	19	11102734	Familial hypercholesterolemia
NM_000527.4(LDLR):c.373C>T (p.Gln125Ter)	LDLR	19	11105279	Familial hypercholesterolemia
NM_000527.4(LDLR):c.418G>A (p.Glu140Lys)	LDLR	19	11105324	Familial hypercholesterolemia
NM_000527.4(LDLR):c.460C>T (p.Gln154Ter)	LDLR	19	11105366	Familial hypercholesterolemia
NM_000527.4(LDLR):c.495G>A (p.Trp165Ter)	LDLR	19	11105401	Familial hypercholesterolemia
NM_000527.4(LDLR):c.539G>A (p.Trp180Ter)	LDLR	19	11105445	Familial hypercholesterolemia
NM_000527.4(LDLR):c.544C>T (p.Gln182Ter)	LDLR	19	11105450	Familial hypercholesterolemia
NM_000527.4(LDLR):c.571C>T (p.Gln191Ter)	LDLR	19	11105477	Familial hypercholesterolemia
NM_000527.4(LDLR):c.642G>A (p.Trp214Ter)	LDLR	19	11105548	Familial hypercholesterolemia
NM_000527.4(LDLR):c.724C>T (p.Gln242Ter)	LDLR	19	11106594	Familial hypercholesterolemia
NM_000527.4(LDLR):c.760C>T (p.Gln254Ter)	LDLR	19	11106630	Familial hypercholesterolemia
NM_000527.4(LDLR):c.817+1G>A	LDLR	19	11106688	Familial hypercholesterolemia
NM_000527.4(LDLR):c.914G>A (p.Trp305Ter)	LDLR	19	11107488	Familial hypercholesterolemia
NM_000527.4(LDLR):c.915G>A (p.Trp305Ter)	LDLR	19	11107489	Familial hypercholesterolemia
NM_000527.4(LDLR):c.941-12G>A	LDLR	19	11110640	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1045C>T (p.Gln349Ter)	LDLR	19	11110756	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1060+1G>A	LDLR	19	11110772	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1061-1G>A	LDLR	19	11111513	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1069G>A (p.Glu357Lys)	LDLR	19	11111522	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1075C>T (p.Gln359Ter)	LDLR	19	11111528	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1096C>T (p.Gln366Ter)	LDLR	19	11111549	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1132C>T (p.Gln378Ter)	LDLR	19	11111585	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1150C>T (p.Gln384Ter)	LDLR	19	11111603	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1186+5G>A	LDLR	19	11111644	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1328G>A (p.Trp443Ter)	LDLR	19	11113419	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1329G>A (p.Trp443Ter)	LDLR	19	11113420	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1342C>T (p.Gln448Ter)	LDLR	19	11113433	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1358+1G>A	LDLR	19	11113450	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1363C>T (p.Gln455Ter)	LDLR	19	11113539	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1420C>T (p.Gln474Ter)	LDLR	19	11113596	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1449G>A (p.Trp483Ter)	LDLR	19	11113625	Familial hypercholesterolemia

NM_000527.4(LDLR):c.1470G>A (p.Trp490Ter)	LDLR	19	11113646	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1586+1G>A	LDLR	19	11113763	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1599G>A (p.Trp533Ter)	LDLR	19	11116106	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1607G>A (p.Trp536Ter)	LDLR	19	11116114	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1633G>A (p.Gly545Arg)	LDLR	19	11116140	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1681C>T (p.Gln561Ter)	LDLR	19	11116188	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1686G>A (p.Trp562Ter)	LDLR	19	11116193	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1735G>A (p.Asp579Asn)	LDLR	19	11116888	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1813C>T (p.Leu605=)	LDLR	19	11116966	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1845+1G>A	LDLR	19	11116999	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1846-1G>A	LDLR	19	11120091	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1859G>A (p.Trp620Ter)	LDLR	19	11120105	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1860G>A (p.Trp620Ter)	LDLR	19	11120106	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1997G>A (p.Trp666Ter)	LDLR	19	11120379	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1998G>A (p.Trp666Ter)	LDLR	19	11120380	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2032C>T (p.Gln678Ter)	LDLR	19	11120414	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2053C>T (p.Pro685Ser)	LDLR	19	11120435	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2056C>T (p.Gln686Ter)	LDLR	19	11120438	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2164C>T (p.Gln722Ter)	LDLR	19	11123197	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2308C>T (p.Gln770Ter)	LDLR	19	11123341	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2311+1G>A	LDLR	19	11123345	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2390-1G>A	LDLR	19	11129512	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2438G>A (p.Trp813Ter)	LDLR	19	11129561	Familial hypercholesterolemia
NM_000527.4(LDLR):c.2547+1G>A	LDLR	19	11129671	Familial hypercholesterolemia
NM_000527.4(LDLR):c.1730G>A (p.Trp577Ter)	LDLR	19	11116883	Familial hypercholesterolemia
NM_000527.4(LDLR):c.910G>A (p.Asp304Asn)	LDLR	19	11107484	Familial hypercholesterolemia;Hypercholesterolaemia
NM_000527.4(LDLR):c.1359-1G>A	LDLR	19	11113534	Familial hypercholesterolemia;Hypercholesterolaemia
NM_000527.4(LDLR):c.301G>A (p.Glu101Lys)	LDLR	19	11102774	Familial hypercholesterolemia;Hypercholesterolaemia;not provided
NM_000527.4(LDLR):c.1775G>A (p.Gly592Glu)	LDLR	19	11116928	Familial hypercholesterolemia;Hypercholesterolaemia;not provided
NM_000384.2(APOB):c.10579C>T (p.Arg3527Trp)	APOB	2	21006289	Familial hypercholesterolemia;Hypercholesterolemia, autosomal dominant, type B
NM_000527.4(LDLR):c.530C>T (p.Ser177Leu)	LDLR	19	11105436	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.2000G>A (p.Cys667Tyr)	LDLR	19	11120382	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.1567G>A (p.Val523Met)	LDLR	19	11113743	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.1646G>A (p.Gly549Asp)	LDLR	19	11116153	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.2054C>T (p.Pro685Leu)	LDLR	19	11120436	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.1055G>A (p.Cys352Tyr)	LDLR	19	11110766	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.502G>A (p.Asp168Asn)	LDLR	19	11105408	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.1246C>T (p.Arg416Trp)	LDLR	19	11113337	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.1784G>A (p.Arg595Gln)	LDLR	19	11116937	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.1897C>T (p.Arg633Cys)	LDLR	19	11120143	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.1247G>A (p.Arg416Gln)	LDLR	19	11113338	Familial hypercholesterolemia;not provided
NM_000527.4(LDLR):c.2430G>A (p.Trp810Ter)	LDLR	19	11129553	Familial hypercholesterolemia;not provided
NM_000352.4(ABCC8):c.4306C>T (p.Arg1436Ter)	ABCC8	11	17395611	Familial hyperinsulinism;Persistent hyperinsulinemic hypoglycemia of infancy
NM_000334.4(SCN4A):c.2111C>T (p.Thr704Met)	SCN4A	17	63957427	Familial hyperkalemic periodic paralysis;Hyperkalemic Periodic Paralysis Type 1;Paramyotonia congenita/hyperkalemic periodic paralysis;not provided

NM_000257.3(MYH7):c.2803G>A (p.Glu935Lys)	MYH7	14	23424026	Familial hypertrophic cardiomyopathy 1
NM_000257.3(MYH7):c.1447G>A (p.Glu483Lys)	MYH7	14	23428631	Familial hypertrophic cardiomyopathy 1
NM_000257.3(MYH7):c.1357C>T (p.Arg453Cys)	MYH7	14	23429005	Familial hypertrophic cardiomyopathy 1;Familial restrictive cardiomyopathy;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.1208G>A (p.Arg403Gln)	MYH7	14	23429278	Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.746G>A (p.Arg249Gln)	MYH7	14	23431468	Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.1207C>T (p.Arg403Trp)	MYH7	14	23429279	Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.2609G>A (p.Arg870His)	MYH7	14	23424839	Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.2389G>A (p.Ala797Thr)	MYH7	14	23425316	Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.2146G>A (p.Gly716Arg)	MYH7	14	23425980	Familial hypertrophic cardiomyopathy 1;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided;not specified
NM_000257.3(MYH7):c.2191C>T (p.Pro731Ser)	MYH7	14	23425790	Familial hypertrophic cardiomyopathy 1;not provided
NM_000257.3(MYH7):c.4135G>A (p.Ala1379Thr)	MYH7	14	23418244	Familial hypertrophic cardiomyopathy 1;Primary familial hypertrophic cardiomyopathy
NM_000257.3(MYH7):c.767G>A (p.Gly256Glu)	MYH7	14	23431447	Familial hypertrophic cardiomyopathy 1;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.5134C>T (p.Arg1712Trp)	MYH7	14	23415652	Familial hypertrophic cardiomyopathy 1;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.2905C>T (p.Gln969Ter)	MYBPC3	11	47335042	Familial hypertrophic cardiomyopathy 1;Primary familial hypertrophic cardiomyopathy;not provided
NM_000432.3(MYL2):c.173G>A (p.Arg58Gln)	MYL2	12	110914287	Familial hypertrophic cardiomyopathy 10;Primary familial hypertrophic cardiomyopathy;not provided
NM_005159.4(ACTC1):c.301G>A (p.Glu101Lys)	ACTC1	15	34793398	Familial hypertrophic cardiomyopathy 11;Left ventricular noncompaction 4;Primary familial hypertrophic cardiomyopathy;not provided
NM_003280.2(TNNC1):c.251G>A (p.Cys84Tyr)	TNNC1	3	52451810	Familial hypertrophic cardiomyopathy 13
NM_002471.3(MYH6):c.2384G>A (p.Arg795Gln)	MYH6	14	23396329	Familial hypertrophic cardiomyopathy 14
NM_020433.4(JPH2):c.494C>T (p.Ser165Phe)	JPH2	20	44160293	Familial hypertrophic cardiomyopathy 17
NM_001001430.2(TNNT2):c.536C>T (p.Ser179Phe)	TNNT2	1	201363330	Familial hypertrophic cardiomyopathy 2;Primary familial hypertrophic cardiomyopathy
NM_001001430.2(TNNT2):c.275G>A (p.Arg92Gln)	TNNT2	1	201365297	Familial hypertrophic cardiomyopathy 2;Primary familial hypertrophic cardiomyopathy;not provided
NM_000364.3(TNNT2):c.304C>T (p.Arg102Trp)	TNNT2	1	201365298	Familial hypertrophic cardiomyopathy 2;Primary familial hypertrophic cardiomyopathy;not provided
NM_001001430.2(TNNT2):c.281G>A (p.Arg94His)	TNNT2	1	201365291	Familial hypertrophic cardiomyopathy 2;Primary familial hypertrophic cardiomyopathy;not provided
NM_001001430.2(TNNT2):c.860G>A (p.Trp287Ter)	TNNT2	1	201359217	Familial hypertrophic cardiomyopathy 2;Primary familial hypertrophic cardiomyopathy;not provided
NM_001018005.1(TPM1):c.523G>A (p.Asp175Asn)	TPM1	15	63060899	Familial hypertrophic cardiomyopathy 3;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000265.5(NCF1):c.*154+25G>A	NCF1	7	74789339	Familial hypertrophic cardiomyopathy 4
NM_000256.3(MYBPC3):c.2065C>T (p.Gln689Ter)	MYBPC3	11	47339653	Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.1484G>A (p.Arg495Gln)	MYBPC3	11	47342718	Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy;MYBPC3-Related Disorders;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3642G>A (p.Trp1214Ter)	MYBPC3	11	47332244	Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.2308+1G>A	MYBPC3	11	47338519	Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3697C>T (p.Gln1233Ter)	MYBPC3	11	47332189	Familial hypertrophic cardiomyopathy 4;Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3330+5G>A	MYBPC3	11	47333189	Familial hypertrophic cardiomyopathy 4;not provided
NM_000256.3(MYBPC3):c.3490+1G>A	MYBPC3	11	47332813	Familial hypertrophic cardiomyopathy 4;Primary familial hypertrophic cardiomyopathy

NM_000256.3(MYBPC3):c.821+5G>A	MYBPC3	11	47347852	Familial hypertrophic cardiomyopathy 4;Primary familial hypertrophic cardiomyopathy	
NM_016203.3(PRKAG2):c.905G>A (p.Arg302Gln)	PRKAG2	7	151576412	Familial hypertrophic cardiomyopathy 6;Glycogen storage disease of heart, lethal congenital;Primary familial hypertrophic cardiomyopathy;Wolff-Parkinson-White pattern;not provided	
NM_000363.4(TNNI3):c.607G>A (p.Gly203Ser)	TNNI3	19	55151860	Familial hypertrophic cardiomyopathy 7	
NM_000363.4(TNNI3):c.61C>T (p.Arg21Cys)	TNNI3	19	55157097	Familial hypertrophic cardiomyopathy 7	
NM_000384.2(APOB):c.3997C>T (p.Arg1333Ter)	APOB	2	21013379	Familial hypobetalipoproteinemia	
NM_000384.2(APOB):c.6253C>T (p.Arg2085Ter)	APOB	2	21010615	Familial hypobetalipoproteinemia	
NM_000384.2(APOB):c.4429C>T (p.Gln1477Ter)	APOB	2	21012439	Familial hypobetalipoproteinemia	
NM_000384.2(APOB):c.7564C>T (p.Arg2522Ter)	APOB	2	21009304	Familial hypobetalipoproteinemia	
NM_000388.3(CASR):c.1525G>A (p.Gly509Arg)	CASR	3	122275959	Familial hypocalciuric hypercalcemia	
NM_000339.2(SLC12A3):c.625C>T (p.Arg209Trp)	SLC12A3	16	56870119	Familial hypokalemia-hypomagnesemia	
NM_000339.2(SLC12A3):c.1964G>A (p.Arg655His)	SLC12A3	16	56886402	Familial hypokalemia-hypomagnesemia	
NM_000339.2(SLC12A3):c.1763C>T (p.Ala588Val)	SLC12A3	16	56884142	Familial hypokalemia-hypomagnesemia	
NM_000339.2(SLC12A3):c.1046C>T (p.Pro349Leu)	SLC12A3	16	56872737	Familial hypokalemia-hypomagnesemia	
NM_000339.2(SLC12A3):c.179C>T (p.Thr60Met)	SLC12A3	16	56865414	Familial hypokalemia-hypomagnesemia	
NM_000339.2(SLC12A3):c.1925G>A (p.Arg642His)	SLC12A3	16	56885364	Familial hypokalemia-hypomagnesemia	
NM_000339.2(SLC12A3):c.2891G>A (p.Arg964Gln)	SLC12A3	16	56904402	Familial hypokalemia-hypomagnesemia	
NM_000458.3(HNF1B):c.529C>T (p.Arg177Ter)	HNF1B	17	37739455	Familial hypoplastic, glomerulocystic kidney	
NM_000458.3(HNF1B):c.826C>T (p.Arg276Ter)	HNF1B	17	37731814	Familial hypoplastic, glomerulocystic kidney	
NM_000458.3(HNF1B):c.494G>A (p.Arg165His)	HNF1B	17	37739490	Familial hypoplastic, glomerulocystic kidney	
NM_020549.4(CHAT):c.1321G>A (p.Glu441Lys)	CHAT	10	49648546	Familial infantile myasthenia	
NM_020549.4(CHAT):c.1679G>A (p.Arg560His)	CHAT	10	49655139	Familial infantile myasthenia	
NM_020549.4(CHAT):c.1493C>T (p.Ser498Leu)	CHAT	10	49649618	Familial infantile myasthenia	
NM_020549.4(CHAT):c.1258C>T (p.Arg420Cys)	CHAT	10	49646651	Familial infantile myasthenia	
NM_020549.4(CHAT):c.418C>T (p.Gln140Ter)	CHAT	10	49619755	Familial infantile myasthenia	
NM_020984.3(CHAT):c.266G>A (p.Arg89His)	CHAT	10	49620535	Familial infantile myasthenia	
NM_003361.3(UMOD):c.443G>A (p.Cys148Tyr)	UMOD	16	20348858	Familial juvenile gout	
NM_003361.3(UMOD):c.230G>A (p.Cys77Tyr)	UMOD	16	20349071	Familial juvenile gout	
NM_003361.3(UMOD):c.764G>A (p.Cys255Tyr)	UMOD	16	20348537	Familial juvenile gout	
NM_000243.2(MEFV):c.800C>T (p.Thr267Ile)	MEFV	16	3254268	Familial Mediterranean fever	
NM_000243.2(MEFV):c.1432C>T (p.His478Tyr)	MEFV	16	3247171	Familial Mediterranean fever;Familial mediterranean fever, autosomal dominant	
NM_000243.2(MEFV):c.2082G>A (p.Met694Ile)	MEFV	16	3243405	Familial Mediterranean fever;Familial mediterranean fever, autosomal dominant;not provided	
NM_000243.2(MEFV):c.2282G>A (p.Arg761His)	MEFV	16	3243205	Familial Mediterranean fever;not provided	
NM_000243.2(MEFV):c.2040G>A (p.Met680Ile)	MEFV	16	3243447	Familial Mediterranean fever;not provided	
NM_000243.2(MEFV):c.1958G>A (p.Arg653His)	MEFV	16	3243529	Familial Mediterranean fever;not provided	
NM_000243.2(MEFV):c.2060G>A (p.Gly687Asp)	MEFV	16	3243427	Familial Mediterranean fever;not provided	
NM_020975.4(RET):c.2410G>A (p.Val804Met)	RET	10	43119548	Familial medullary thyroid carcinoma;Hereditary cancer-predisposing syndrome;Multiple endocrine neoplasia, type 2;Multiple endocrine neoplasia, type 2b;not provided	
NM_005572.3(LMNA):c.1444C>T (p.Arg482Trp)	LMNA	1	156136984	Familial partial lipodystrophy 2;not provided	
NM_170707.3(LMNA):c.1394G>A (p.Gly465Asp)	LMNA	1	156136934	Familial partial lipodystrophy 2;not provided	
NM_170707.3(LMNA):c.1745G>A (p.Arg582His)	LMNA	1	156138534	Familial partial lipodystrophy 2;not provided	
NM_170707.3(LMNA):c.688G>A (p.Asp230Asn)	LMNA	1	156134853	Familial partial lipodystrophy 2;not provided	
NM_138712.3(PPARG):c.868G>A (p.Val290Met)	PPARG	3	12416836	Familial partial lipodystrophy 3	
NM_138712.3(PPARG):c.1189C>T (p.Arg397Cys)	PPARG	3	12433900	Familial partial lipodystrophy 3	
NM_138712.3(PPARG):c.496C>T (p.Arg166Trp)	PPARG	3	12392713	Familial partial lipodystrophy 3	

NM_001754.4(RUNX1):c.602G>A (p.Arg201Gln)	RUNX1	21	34859485	Familial platelet disorder with associated myeloid malignancy	
NM_000374.4(UROD):c.942G>A (p.Glu314=)	UROD	1	45015006	Familial porphyria cutanea tarda	
NM_000374.4(UROD):c.583C>T (p.Leu195Phe)	UROD	1	45014017	Familial porphyria cutanea tarda	
NM_000374.4(UROD):c.995G>A (p.Arg332His)	UROD	1	45015389	Familial porphyria cutanea tarda	
NM_000374.4(UROD):c.842G>A (p.Gly281Glu)	UROD	1	45014803	Familial porphyria cutanea tarda;Hepatoerythropoietic porphyria;Porphyria cutanea tarda	
NM_001013703.3(EIF2AK4):c.3802C>T (p.Gln1268Ter)	EIF2AK4	15	40016544	Familial pulmonary capillary hemangiomatosis	
NM_001013703.3(EIF2AK4):c.3406C>T (p.Arg1136Ter)	EIF2AK4	15	40007064	Familial pulmonary capillary hemangiomatosis	
NM_001013703.3(EIF2AK4):c.1754G>A (p.Arg585Gln)	EIF2AK4	15	39973685	Familial pulmonary capillary hemangiomatosis	
NM_001013703.3(EIF2AK4):c.1387C>T (p.Arg463Ter)	EIF2AK4	15	39967713	Familial pulmonary capillary hemangiomatosis	
NM_001013703.3(EIF2AK4):c.3766C>T (p.Arg1256Ter)	EIF2AK4	15	40016508	Familial pulmonary capillary hemangiomatosis	
NM_001013703.3(EIF2AK4):c.3448C>T (p.Arg1150Ter)	EIF2AK4	15	40008067	Familial pulmonary capillary hemangiomatosis	
NM_001013703.3(EIF2AK4):c.860-1G>A	EIF2AK4	15	39965685	Familial pulmonary capillary hemangiomatosis	
NM_003041.3(SLC5A2):c.1320G>A (p.Trp440Ter)	SLC5A2	16	31488919	Familial renal glucosuria	
NM_003041.3(SLC5A2):c.265G>A (p.Ala89Thr)	SLC5A2	16	31484885	Familial renal glucosuria	
NM_144585.3(SLC22A12):c.774G>A (p.Trp258Ter)	SLC22A12	11	64593747	Familial renal hypouricemia	
NM_144585.3(SLC22A12):c.650C>T (p.Thr217Met)	SLC22A12	11	64593548	Familial renal hypouricemia	
NM_144585.3(SLC22A12):c.269G>A (p.Arg90His)	SLC22A12	11	64591825	Familial renal hypouricemia	
NM_000363.4(TNNI3):c.433C>T (p.Arg145Trp)	TNNI3	19	55154146	Familial restrictive cardiomyopathy 1;Primary familial hypertrophic cardiomyopathy;not provided	
NM_001001430.2(TNNT2):c.280C>T (p.Arg94Cys)	TNNT2	1	201365292	Familial restrictive cardiomyopathy;not provided	
NM_000041.3(APOE):c.487C>T (p.Arg163Cys)	APOE	19	44908783	Familial type 3 hyperlipoproteinemia	
NM_000041.3(APOE):c.683G>A (p.Trp228Ter)	APOE	19	44908979	Familial type 3 hyperlipoproteinemia	
NM_000041.3(APOE):c.488G>A (p.Arg163His)	APOE	19	44908784	Familial type 3 hyperlipoproteinemia	
NM_052968.4(APOA5):c.415C>T (p.Gln139Ter)	APOA5	11	116790814	Familial type 5 hyperlipoproteinemia	
NM_004048.2(B2M):c.286G>A (p.Asp96Asn)	B2M	15	44715641	Familial visceral amyloidosis, Ostertag type	
NM_000444.5(PHEX):c.254G>A (p.Cys85Tyr)	PHEX	X	22047116	Familial X-linked hypophosphatemic vitamin D refractory rickets	
NM_000444.5(PHEX):c.1699C>T (p.Arg567Ter)	PHEX	X	22212957	Familial X-linked hypophosphatemic vitamin D refractory rickets;not provided	
NM_000444.5(PHEX):c.1735G>A (p.Gly579Arg)	PHEX	X	22219070	Familial X-linked hypophosphatemic vitamin D refractory rickets;not provided	
NM_033084.4(FANCD2):c.2444G>A (p.Arg815Gln)	FANCD2	3	10067267	Fanconi anemia	
NM_001113378.1(FANCI):c.507G>A (p.Trp169Ter)	FANCI	15	89263422	Fanconi anemia	
NM_020937.3(FANCM):c.5101C>T (p.Gln1701Ter)	FANCM	14	45189123	Fanconi anemia	
NM_000135.2(FANCA):c.2851C>T (p.Arg951Trp)	FANCA	16	89761950	Fanconi anemia	
NM_000135.2(FANCA):c.2749C>T (p.Arg917Trp)	FANCA	16	89764919	Fanconi anemia	
NM_000135.2(FANCA):c.1827-1G>A	FANCA	16	89775816	Fanconi anemia	
NM_000135.2(FANCA):c.709+5G>A	FANCA	16	89805275	Fanconi anemia	
NM_000135.2(FANCA):c.513G>A (p.Trp171Ter)	FANCA	16	89810716	Fanconi anemia, complementation group A	
NM_000135.2(FANCA):c.2557C>T (p.Arg853Ter)	FANCA	16	89767185	Fanconi anemia, complementation group A	
NM_000135.2(FANCA):c.295C>T (p.Gln99Ter)	FANCA	16	89811060	Fanconi anemia, complementation group A	
NM_000135.2(FANCA):c.154C>T (p.Arg52Ter)	FANCA	16	89815912	Fanconi anemia, complementation group A	
NM_001018115.2(FANCD2):c.3707G>A (p.Arg1236His)	FANCD2	3	10090315	Fanconi anemia, complementation group D2	
NM_001018115.2(FANCD2):c.904C>T (p.Arg302Trp)	FANCD2	3	10043065	Fanconi anemia, complementation group D2	
NM_001018115.2(FANCD2):c.958C>T (p.Gln320Ter)	FANCD2	3	10043119	Fanconi anemia, complementation group D2	
NM_021922.2(FANCE):c.355C>T (p.Gln119Ter)	FANCE	6	35455853	Fanconi anemia, complementation group E	
NM_021922.2(FANCE):c.421C>T (p.Arg141Ter)	FANCE	6	35455919	Fanconi anemia, complementation group E	
NG_011708.1:g.11963G>A	FANCE	6	35459323	Fanconi anemia, complementation group E	
NM_022725.3(FANCF):c.16C>T (p.Gln6Ter)	FANCF	11	22625795	Fanconi anemia, complementation group F	

NM_004629.1(FANCG):c.1066C>T (p.Gln356Ter)	FANCG	9	35076442	Fanconi anemia, complementation group G	
NM_001113378.1(FANCI):c.3854G>A (p.Arg1285Gln)	FANCI	15	89315319	Fanconi anemia, complementation group I	
NM_001113378.1(FANCI):c.3853C>T (p.Arg1285Ter)	FANCI	15	89315318	Fanconi anemia, complementation group I	
NM_032043.2(BRIP1):c.1240C>T (p.Gln414Ter)	BRIP1	17	61799200	Fanconi anemia, complementation group J;Hereditary cancer-predisposing syndrome;Neoplasm of ovary;not provided	
NM_058216.2(RAD51C):c.773G>A (p.Arg258His)	RAD51C	17	58709926	Fanconi anemia, complementation group O;Hereditary cancer-predisposing syndrome	
NM_058216.2(RAD51C):c.955C>T (p.Arg319Ter)	RAD51C	17	58724090	Fanconi anemia, complementation group O;Hereditary cancer-predisposing syndrome;not provided	
NM_058216.2(RAD51C):c.577C>T (p.Arg193Ter)	RAD51C	17	58703201	Fanconi anemia, complementation group O;Hereditary cancer-predisposing syndrome;not provided	
NM_058216.2(RAD51C):c.709C>T (p.Arg237Ter)	RAD51C	17	58709862	Fanconi anemia, complementation group O;Hereditary cancer-predisposing syndrome;not provided	
NM_002875.4(RAD51):c.877G>A (p.Ala293Thr)	RAD51	15	40729955	FANCONI ANEMIA, COMPLEMENTATION GROUP R	
NM_014176.3(UBE2T):c.179+5G>A	UBE2T	1	202334984	Fanconi anemia, complementation group t	
NM_000136.2(FANCC):c.339G>A (p.Trp113Ter)	FANCC	9	95240655	Fanconi anemia;Fanconi anemia, complementation group C	
NM_000136.2(FANCC):c.553C>T (p.Arg185Ter)	FANCC	9	95150056	Fanconi anemia;Fanconi anemia, complementation group C;not provided	
NM_000136.2(FANCC):c.37C>T (p.Gln13Ter)	FANCC	9	95249255	Fanconi anemia;Fanconi anemia, complementation group C;not provided	
NM_000136.2(FANCC):c.1642C>T (p.Arg548Ter)	FANCC	9	95101742	Fanconi anemia;Fanconi anemia, complementation group C;not provided	
NM_000136.2(FANCC):c.319C>T (p.Gln107Ter)	FANCC	9	95240675	Fanconi anemia;not provided	
NM_001966.3(EHHADH):c.7G>A (p.Glu3Lys)	EHHADH	3	185254016	Fanconi renotubular syndrome 3	
NM_000457.4(HNF4A):c.253C>T (p.Arg85Trp)	HNF4A	20	44406195	Fanconi renotubular syndrome 4 with maturity-onset diabetes of the young;Hyperinsulinemia;not provided	
NM_000340.1(SLC2A2):c.1093C>T (p.Arg365Ter)	SLC2A2	3	170999142	Fanconi-Bickel syndrome	
NM_000340.1(SLC2A2):c.901C>T (p.Arg301Ter)	SLC2A2	3	171005347	Fanconi-Bickel syndrome	
NM_000340.1(SLC2A2):c.1250C>T (p.Pro417Leu)	SLC2A2	3	170998317	Fanconi-Bickel syndrome	
NM_000340.1(SLC2A2):c.1259G>A (p.Trp420Ter)	SLC2A2	3	170998308	Fanconi-Bickel syndrome	
NM_000340.1(SLC2A2):c.859C>T (p.Gln287Ter)	SLC2A2	3	171005389	Fanconi-Bickel syndrome	
NM_000311.4(PRNP):c.598G>A (p.Glu200Lys)	PRNP	20	4699818	Fatal familial insomnia;Genetic prion diseases;Jakob-Creutzfeldt disease	
NM_000311.4(PRNP):c.532G>A (p.Asp178Asn)	PRNP	20	4699752	Fatal familial insomnia;Genetic prion diseases;Jakob-Creutzfeldt disease	
NM_002473.5(MYH9):c.3464C>T (p.Thr1155Ile)	MYH9	22	36295526	Fechtner syndrome;May-Hegglin anomaly	
NM_002473.5(MYH9):c.4270G>A (p.Asp1424Asn)	MYH9	22	36292060	Fechtner syndrome;MYH9 related disorders;Macrothrombocytopenia and progressive sensorineural deafness;May-Hegglin anomaly;Sebastian syndrome	
NM_002473.5(MYH9):c.5521G>A (p.Glu1841Lys)	MYH9	22	36284474	Fechtner syndrome;MYH9 related disorders;May-Hegglin anomaly	
NM_002473.5(MYH9):c.5797C>T (p.Arg1933Ter)	MYH9	22	36282754	Fechtner syndrome;MYH9 related disorders;May-Hegglin anomaly;Sebastian syndrome	
NM_005378.5(MYCN):c.1178G>A (p.Arg393His)	MYCN	2	15945880	Feingold syndrome 1	
NM_005378.5(MYCN):c.1181G>A (p.Arg394His)	MYCN	2	15945883	Feingold syndrome 1	
NM_005378.5(MYCN):c.231G>A (p.Trp77Ter)	MYCN	2	15942295	Feingold syndrome 1	
NM_005378.5(MYCN):c.1145G>A (p.Arg382His)	MYCN	2	15945847	Feingold syndrome 1	
NM_000184.2(HBG2):c.-167C>T	HBG2	11	5254895	Fetal hemoglobin quantitative trait locus 1	
NM_000559.2(HBG1):c.-53-196C>T	HBG1	11	5250053	Fetal hemoglobin quantitative trait locus 1	
NM_000559.2(HBG1):c.-167C>T	HBG1	11	5249971	Fetal hemoglobin quantitative trait locus 1	
NM_000559.2(HBG1):c.-170G>A	HBG1	11	5249974	Fetal hemoglobin quantitative trait locus 1;Greek HPFH;Sardinian HPFH	
NM_005120.2(MED12):c.2873G>A (p.Gly958Glu)	MED12	X	71127359	FG syndrome	
NM_005120.2(MED12):c.2444G>A (p.Arg815Gln)	MED12	X	71126057	FG syndrome	
NM_001110556.1(FLNA):c.3872C>T (p.Pro1291Leu)	FLNA	X	154359839	FG syndrome 2	
NM_003688.3(CASK):c.1186C>T (p.Pro396Ser)	CASK	X	41589562	FG syndrome 4	
NM_003688.3(CASK):c.79C>T (p.Arg27Ter)	CASK	X	41853208	FG syndrome 4;Mental retardation and microcephaly with pontine and cerebellar hypoplasia	
NM_005120.2(MED12):c.4147G>A (p.Ala1383Thr)	MED12	X	71132100	FG syndrome;not provided	

NM_021870.2(FGG):c.571G>A (p.Gly191Arg)	FGG	4	154609725	Fibrinogen Milano XII, digenic
NM_001173464.1(KIF21A):c.2861G>A (p.Arg954Gln)	KIF21A	12	39332404	Fibrosis of extraocular muscles, congenital, 1;Fibrosis of extraocular muscles, congenital, 3b
NM_001173464.1(KIF21A):c.2860C>T (p.Arg954Trp)	KIF21A	12	39332405	Fibrosis of extraocular muscles, congenital, 1;Fibrosis of extraocular muscles, congenital, 3b;not provided
NM_005169.3(PHOX2A):c.215C>T (p.Ala72Val)	PHOX2A	11	72243790	Fibrosis of extraocular muscles, congenital, 2
NM_006086.3(TUBB3):c.1249G>A (p.Asp417Asn)	TUBB3	16	89935700	Fibrosis of extraocular muscles, congenital, 3a, with or without extraocular involvement
NM_006086.3(TUBB3):c.185G>A (p.Arg62Gln)	TUBB3	16	89933486	Fibrosis of extraocular muscles, congenital, 3a, with or without extraocular involvement
NM_006086.3(TUBB3):c.785G>A (p.Arg262His)	TUBB3	16	89935236	Fibrosis of extraocular muscles, congenital, 3a, with or without extraocular involvement
NM_006086.3(TUBB3):c.784C>T (p.Arg262Cys)	TUBB3	16	89935235	Fibrosis of extraocular muscles, congenital, 3a, with or without extraocular involvement;not provided
NM_006086.3(TUBB3):c.904G>A (p.Ala302Thr)	TUBB3	16	89935355	Fibrosis of extraocular muscles, congenital, 3a, with or without extraocular involvement;not provided
NM_006086.3(TUBB3):c.211G>A (p.Gly71Arg)	TUBB3	16	89933512	Fibrosis of extraocular muscles, congenital, 3a, with or without extraocular involvement;not provided
NM_006086.3(TUBB3):c.1138C>T (p.Arg380Cys)	TUBB3	16	89935589	Fibrosis of extraocular muscles, congenital, 3a, with or without extraocular involvement;not provided
NM_001173464.1(KIF21A):c.2841G>A (p.Met947Ile)	KIF21A	12	39332606	Fibrosis of extraocular muscles, congenital, 3b
NM_198721.3(COL25A1):c.1144G>A (p.Gly382Arg)	COL25A1	4	108863327	Fibrosis of extraocular muscles, congenital, 5
NM_001122681.1(SH3BP2):c.1253C>T (p.Pro418Leu)	SH3BP2	4	2831582	Fibrous dysplasia of jaw
NM_001122681.1(SH3BP2):c.1244G>A (p.Arg415Gln)	SH3BP2	4	2831573	Fibrous dysplasia of jaw
NM_001122681.1(SH3BP2):c.1259G>A (p.Gly420Glu)	SH3BP2	4	2831588	Fibrous dysplasia of jaw
NM_000557.4(GDF5):c.1133G>A (p.Arg378Gln)	GDF5	20	35434282	Fibular hypoplasia and complex brachydactyly
NM_000094.3(COL7A1):c.2005C>T (p.Arg669Ter)	COL7A1	3	48590258	Finger syndactyly;Palmoplantar blistering;Short stature;Toe syndactyly;not provided
NM_004646.3(NPHS1):c.3325C>T (p.Arg1109Ter)	NPHS1	19	35831358	Finnish congenital nephrotic syndrome
NM_004646.3(NPHS1):c.3478C>T (p.Arg1160Ter)	NPHS1	19	35831056	Finnish congenital nephrotic syndrome
NM_004646.3(NPHS1):c.2464G>A (p.Val822Met)	NPHS1	19	35842421	Finnish congenital nephrotic syndrome
NM_004646.3(NPHS1):c.532C>T (p.Gln178Ter)	NPHS1	19	35850440	Finnish congenital nephrotic syndrome
NM_004646.3(NPHS1):c.2335-1G>A	NPHS1	19	35842551	Finnish congenital nephrotic syndrome
NM_004646.3(NPHS1):c.1715G>A (p.Ser572Asn)	NPHS1	19	35845711	Finnish congenital nephrotic syndrome;not provided
NM_000229.1(LCAT):c.440C>T (p.Thr147Ile)	LCAT	16	67942754	Fish-eye disease
NM_000229.1(LCAT):c.101C>T (p.Pro34Leu)	LCAT	16	67944001	Fish-eye disease
NM_000229.1(LCAT):c.1112C>T (p.Thr371Met)	LCAT	16	67940115	Fish-eye disease
NM_024665.5(TBL1XR1):c.209G>A (p.Gly70Asp)	TBL1XR1	3	177051722	Fitzsimmons-Guilbert syndrome
NM_015040.3(PIKFYVE):c.2962C>T (p.Gln988Ter)	PIKFYVE	2	208325773	Fleck corneal dystrophy
NM_001457.3(FLNB):c.4835G>A (p.Gly1612Asp)	FLNB	3	58136142	FLNB-Related Disorders
NM_001457.3(FLNB):c.502G>A (p.Gly168Ser)	FLNB	3	58077255	FLNB-Related Disorders
NM_006662.2(SRCAP):c.7000C>T (p.Gln2334Ter)	SRCAP	16	30736616	Floating-Harbor syndrome
NM_006662.2(SRCAP):c.7993C>T (p.Gln2665Ter)	SRCAP	16	30738033	Floating-Harbor syndrome
NM_006662.2(SRCAP):c.7330C>T (p.Arg2444Ter)	SRCAP	16	30737370	Floating-Harbor syndrome;not provided
NM_006662.2(SRCAP):c.7303C>T (p.Arg2435Ter)	SRCAP	16	30737343	Floating-Harbor syndrome;not provided
NM_203473.2(PORCN):c.178G>A (p.Gly60Arg)	PORCN	X	48511336	Focal dermal hypoplasia
NM_203473.2(PORCN):c.370C>T (p.Arg124Ter)	PORCN	X	48511932	Focal dermal hypoplasia
NM_203473.2(PORCN):c.222G>A (p.Trp74Ter)	PORCN	X	48511380	Focal dermal hypoplasia
NM_203473.2(PORCN):c.1094G>A (p.Arg365Gln)	PORCN	X	48516082	Focal dermal hypoplasia
NM_003159.2(CDKL5):c.2641C>T (p.Gln881Ter)	CDKL5	X	18628515	Focal epilepsy
NM_006920.4(SCN1A):c.4753C>T (p.Arg1585Cys)	SCN1A	2	165994212	Focal epilepsy;not provided
NM_004924.5(ACTN4):c.776C>T (p.Thr259Ile)	ACTN4	19	38710299	Focal segmental glomerulosclerosis 1
NM_004924.5(ACTN4):c.584G>A (p.Gly195Asp)	ACTN4	19	38708128	Focal segmental glomerulosclerosis 1
NM_004621.5(TRPC6):c.2683C>T (p.Arg895Cys)	TRPC6	11	101453068	Focal segmental glomerulosclerosis 2

NM_004621.5(TRPC6):c.2689G>A (p.Glu897Lys)	TRPC6	11	101453062	Focal segmental glomerulosclerosis 2
NM_012120.2(CD2AP):c.1834C>T (p.Arg612Ter)	CD2AP	6	47612492	Focal segmental glomerulosclerosis 3
NM_022489.3(INF2):c.653G>A (p.Arg218Gln)	INF2	14	104703440	Focal segmental glomerulosclerosis 5
NM_022489.3(INF2):c.652C>T (p.Arg218Trp)	INF2	14	104703439	Focal segmental glomerulosclerosis 5
NM_022489.3(INF2):c.641G>A (p.Arg214His)	INF2	14	104703428	Focal segmental glomerulosclerosis 5
NM_003990.4(PAX2):c.167G>A (p.Arg56Gln)	PAX2	10	100749869	Focal segmental glomerulosclerosis 7
NM_018685.4(ANLN):c.1291C>T (p.Arg431Cys)	ANLN	7	36411062	Focal segmental glomerulosclerosis 8
NM_173689.6(CRB2):c.1882C>T (p.Arg628Cys)	CRB2	9	123370935	Focal segmental glomerulosclerosis 9
NM_173689.6(CRB2):c.3746G>A (p.Arg1249Gln)	CRB2	9	123376950	Focal segmental glomerulosclerosis 9
NM_001174136.1(TDGF1):c.326C>T (p.Pro109Leu)	TDGF1	3	46579986	Forebrain defects
NM_001080442.2(SLC38A8):c.697G>A (p.Glu233Lys)	SLC38A8	16	84022883	Foveal hypoplasia 2
NM_001080442.2(SLC38A8):c.598C>T (p.Gln200Ter)	SLC38A8	16	84031901	Foveal hypoplasia 2 and optic nerve misrouting with or without anterior segment dysgenesis
NM_000280.4(PAX6):c.382C>T (p.Arg128Cys)	PAX6	11	31800832	Foveal hypoplasia and presenile cataract syndrome
NM_001017995.2(SH3PXD2B):c.127C>T (p.Arg43Trp)	SH3PXD2B	5	172422445	Frank Ter Haar syndrome
NM_002470.3(MYH3):c.2015G>A (p.Arg672His)	MYH3	17	10641317	Freeman-Sheldon syndrome
NM_002470.3(MYH3):c.2014C>T (p.Arg672Cys)	MYH3	17	10641318	Freeman-Sheldon syndrome
NM_001456.3(FLNA):c.3668C>T (p.Pro1223Leu)	FLNA	X	154360127	Frontometaphyseal dysplasia
NM_145331.2(MAP3K7):c.1535C>T (p.Pro512Leu)	MAP3K7	6	90518552	FRONTOMETAPHYSEAL DYSPLASIA 2
NM_001110556.1(FLNA):c.3557C>T (p.Ser1186Leu)	FLNA	X	154360238	Frontometaphyseal dysplasia;not provided
NM_006492.2(ALX3):c.547C>T (p.Arg183Trp)	ALX3	1	110064634	Frontonasal dysplasia 1
NM_006492.2(ALX3):c.586C>T (p.Arg196Trp)	ALX3	1	110064595	Frontonasal dysplasia 1
NM_021926.3(ALX4):c.793C>T (p.Arg265Ter)	ALX4	11	44267607	Frontonasal dysplasia 2
NM_006982.2(ALX1):c.531+1G>A	ALX1	12	85283877	Frontonasal dysplasia 3
NM_002087.3(GRN):c.708+1G>A	GRN	17	44350801	Frontotemporal dementia
NM_001301339.1(CHCHD10):c.239C>T (p.Pro80Leu)	CHCHD10	22	23767396	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2
NM_001301339.1(CHCHD10):c.176C>T (p.Ser59Leu)	CHCHD10	22	23767459	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2;Myopathy, isolated mitochondrial, autosomal dominant
NM_001142298.1(SQSTM1):c.-47-1924C>T	SQSTM1	5	179821034	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3
NM_003900.4(SQSTM1):c.1160C>T (p.Pro387Leu)	SQSTM1	5	179833777	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3;not provided
NM_003900.4(SQSTM1):c.1175C>T (p.Pro392Leu)	SQSTM1	5	179836445	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3;Paget disease of bone;Paget disease of bone, familial
NM_013254.3(TBK1):c.1340+1G>A	TBK1	12	64486018	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4
NM_013254.3(TBK1):c.2086G>A (p.Glu696Lys)	TBK1	12	64497987	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4
NM_014043.3(CHMP2B):c.532-1G>A	CHMP2B	3	87253711	Frontotemporal Dementia, Chromosome 3-Linked
NM_014043.3(CHMP2B):c.493C>T (p.Gln165Ter)	CHMP2B	3	87253472	Frontotemporal Dementia, Chromosome 3-Linked;not provided
NM_014043.3(CHMP2B):c.556C>T (p.Arg186Ter)	CHMP2B	3	87253736	Frontotemporal Dementia, Chromosome 3-Linked;not provided
NM_002087.3(GRN):c.835+1G>A	GRN	17	44351164	Frontotemporal dementia, ubiquitin-positive
NM_002087.3(GRN):c.373C>T (p.Gln125Ter)	GRN	17	44350251	Frontotemporal dementia, ubiquitin-positive;not provided
NM_002087.3(GRN):c.3G>A (p.Met1Ile)	GRN	17	44349167	Frontotemporal dementia, ubiquitin-positive;not provided
NM_002087.3(GRN):c.1477C>T (p.Arg493Ter)	GRN	17	44352404	Frontotemporal dementia, ubiquitin-positive;not provided
NM_016835.4(MAPT):c.2167C>T (p.Arg723Trp)	MAPT	17	46024061	Frontotemporal dementia;not provided
NM_016835.4(MAPT):c.1866+14C>T	MAPT	17	46010416	Frontotemporal dementia;not provided
NM_016835.4(MAPT):c.1960G>A (p.Val654Met)	MAPT	17	46018629	Frontotemporal dementia;not provided
NM_016835.4(MAPT):c.1865G>A (p.Ser622Asn)	MAPT	17	46010401	Frontotemporal dementia;not provided
NM_016835.4(MAPT):c.1852C>T (p.Pro618Ser)	MAPT	17	46010388	Frontotemporal dementia;not provided

NM_016835.4(MAPT):c.1853C>T (p.Pro618Leu)	MAPT	17	46010389	Frontotemporal dementia;Progressive supranuclear ophthalmoplegia;not provided
NM_000507.3(FBP1):c.490G>A (p.Gly164Ser)	FBP1	9	94609998	Fructose-biphosphatase deficiency
NM_000507.3(FBP1):c.825+1G>A	FBP1	9	94605456	Fructose-biphosphatase deficiency
NM_000507.3(FBP1):c.355G>A (p.Asp119Asn)	FBP1	9	94617839	Fructose-biphosphatase deficiency;not provided
NM_000147.4(FUCA1):c.1279C>T (p.Gln427Ter)	FUCA1	1	23845837	Fucosidosis
NM_000147.4(FUCA1):c.244C>T (p.Gln82Ter)	FUCA1	1	23868043	Fucosidosis
NM_000147.4(FUCA1):c.1160G>A (p.Trp387Ter)	FUCA1	1	23848649	Fucosidosis
NM_000147.4(FUCA1):c.464C>T (p.Ser155Phe)	FUCA1	1	23865551	Fucosidosis
NM_000147.4(FUCA1):c.790C>T (p.Arg264Ter)	FUCA1	1	23854539	Fucosidosis
NM_000147.4(FUCA1):c.1125G>A (p.Trp375Ter)	FUCA1	1	23848684	Fucosidosis
NM_000150.2(FUT6):c.739G>A (p.Glu247Lys)	FUT6	19	5831829	Fucosyltransferase 6 deficiency
NM_004625.3(WNT7A):c.325G>A (p.Ala109Thr)	WNT7A	3	13854777	Fuhrmann syndrome
NM_001079802.1(FKTN):c.919C>T (p.Arg307Ter)	FKTN	9	105617967	Fukuyama congenital muscular dystrophy
NM_000143.3(FH):c.793G>A (p.Ala265Thr)	FH	1	241506114	Fumarase deficiency
NM_000143.3(FH):c.267+1G>A	FH	1	241517181	Fumarase deficiency
NM_000143.3(FH):c.679C>T (p.Gln227Ter)	FH	1	241508662	Fumarase deficiency
NM_000143.3(FH):c.698G>A (p.Arg233His)	FH	1	241508643	Fumarase deficiency;Hereditary leiomyomatosis and renal cell cancer;not provided
NM_000143.3(FH):c.1189G>A (p.Gly397Arg)	FH	1	241502490	Fumarase deficiency;Hereditary leiomyomatosis and renal cell cancer;not provided
NM_000143.3(FH):c.1097G>A (p.Ser366Asn)	FH	1	241504053	Fumarase deficiency;not provided
NM_000137.2(FAH):c.1021C>T (p.Arg341Trp)	FAH	15	80180184	Fumarylacetoacetase pseudodeficiency;Tyrosinemia type I;not provided
NM_002905.3(RDH5):c.218C>T (p.Ser73Phe)	RDH5	12	55721402	Fundus albipunctatus, autosomal recessive
NM_002905.3(RDH5):c.839G>A (p.Arg280His)	RDH5	12	55724427	Fundus albipunctatus, autosomal recessive
NM_002905.3(RDH5):c.469C>T (p.Arg157Trp)	RDH5	12	55721847	Fundus albipunctatus, autosomal recessive
NM_000403.3(GALE):c.280G>A (p.Val94Met)	GALE	1	23798188	Galactose epimerase deficiency, severe;UDPglucose-4-epimerase deficiency
NM_000308.3(CTSA):c.1369G>A (p.Gly457Ser)	CTSA	20	45898065	Galactosialidosis, early infantile
NM_000153.3(GALC):c.1630G>A (p.Asp544Asn)	GALC	14	87945593	Galactosylceramide beta-galactosidase deficiency
NM_000153.3(GALC):c.121G>A (p.Gly41Ser)	GALC	14	87993044	Galactosylceramide beta-galactosidase deficiency
NM_000153.3(GALC):c.169G>A (p.Gly57Ser)	GALC	14	87992996	Galactosylceramide beta-galactosidase deficiency
NM_000153.3(GALC):c.205C>T (p.Arg69Ter)	GALC	14	87988514	Galactosylceramide beta-galactosidase deficiency
NM_000153.3(GALC):c.331G>A (p.Gly111Ser)	GALC	14	87986600	Galactosylceramide beta-galactosidase deficiency
NM_000153.3(GALC):c.857G>A (p.Gly286Asp)	GALC	14	87968386	Galactosylceramide beta-galactosidase deficiency;not provided
NM_000153.3(GALC):c.1592G>A (p.Arg531His)	GALC	14	87945631	Galactosylceramide beta-galactosidase deficiency;not provided
NM_032856.3(WDR73):c.703C>T (p.Gln235Ter)	WDR73	15	84645651	Galloway-Mowat syndrome
NM_032856.3(WDR73):c.1039C>T (p.His347Tyr)	WDR73	15	84643568	Galloway-Mowat syndrome
NM_032856.3(WDR73):c.940C>T (p.Gln314Ter)	WDR73	15	84643667	Galloway-Mowat syndrome
NM_032856.3(WDR73):c.287G>A (p.Arg96Lys)	WDR73	15	84648537	Galloway-Mowat syndrome
NM_000663.4(ABAT):c.659G>A (p.Arg220Lys)	ABAT	16	8768248	Gamma-aminobutyric acid transaminase deficiency
NM_000663.4(ABAT):c.275G>A (p.Arg92Gln)	ABAT	16	8750498	Gamma-aminobutyric acid transaminase deficiency
NM_000663.4(ABAT):c.631C>T (p.Leu211Phe)	ABAT	16	8768220	Gamma-aminobutyric acid transaminase deficiency
NM_020533.2(MCOLN1):c.304C>T (p.Arg102Ter)	MCOLN1	19	7526505	Ganglioside sialidase deficiency
NM_020533.2(MCOLN1):c.1207C>T (p.Arg403Cys)	MCOLN1	19	7529173	Ganglioside sialidase deficiency
NM_020533.2(MCOLN1):c.235C>T (p.Gln79Ter)	MCOLN1	19	7525164	Ganglioside sialidase deficiency
NM_020533.2(MCOLN1):c.514C>T (p.Arg172Ter)	MCOLN1	19	7526869	Ganglioside sialidase deficiency
NM_020533.2(MCOLN1):c.1388G>A (p.Cys463Tyr)	MCOLN1	19	7530314	Ganglioside sialidase deficiency
NM_020533.2(MCOLN1):c.964C>T (p.Arg322Ter)	MCOLN1	19	7528683	Ganglioside sialidase deficiency;not provided

NM_000404.3(GLB1):c.1370G>A (p.Arg457Gln)	GLB1	3	33016818	Gangliosidosis GM1 type 3
NM_000404.3(GLB1):c.245C>T (p.Thr82Met)	GLB1	3	33072544	Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis
NM_000038.5(APC):c.622C>T (p.Gln208Ter)	APC	5	112780880	Gardner syndrome
NM_003000.2(SDHB):c.79C>T (p.Arg27Ter)	SDHB	1	17044882	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma
NM_003000.2(SDHB):c.724C>T (p.Arg242Cys)	SDHB	1	17022649	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma
NM_003000.2(SDHB):c.640C>T (p.Gln214Ter)	SDHB	1	17023975	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma
NM_003000.2(SDHB):c.725G>A (p.Arg242His)	SDHB	1	17022648	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma;not provided
NM_003000.2(SDHB):c.423+1G>A	SDHB	1	17028599	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma;not provided
NM_003000.2(SDHB):c.689G>A (p.Arg230His)	SDHB	1	17022684	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma;not provided
NM_003000.2(SDHB):c.136C>T (p.Arg46Ter)	SDHB	1	17044825	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma;not provided
NM_003000.2(SDHB):c.137G>A (p.Arg46Gln)	SDHB	1	17044824	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma;not provided
NM_003000.2(SDHB):c.587G>A (p.Cys196Tyr)	SDHB	1	17024028	Gastrointestinal stromal tumor;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma;not provided
NM_003000.2(SDHB):c.268C>T (p.Arg90Ter)	SDHB	1	17033078	Gastrointestinal stromal tumor;Hereditary Paraganglioma-Pheochromocytoma Syndromes;Hereditary cancer-predisposing syndrome;Paragangliomas 4;Pheochromocytoma;not provided
NM_000222.2(KIT):c.2515G>A (p.Glu839Lys)	KIT	4	54736528	Gastrointestinal stromal tumor;Mastocytosis, sporadic, childhood-onset
NM_003000.2(SDHB):c.602G>A (p.Trp201Ter)	SDHB	1	17024013	Gastrointestinal stromal tumor;Paragangliomas 4;Pheochromocytoma
NM_003000.2(SDHB):c.141G>A (p.Trp47Ter)	SDHB	1	17044820	Gastrointestinal stromal tumor;Paragangliomas 4;Pheochromocytoma
NM_001042465.2(PSAP):c.1297C>T (p.Gln433Ter)	PSAP	10	71819527	Gaucher disease, atypical, due to saposin C deficiency
NM_001005741.2(GBA):c.1192C>T (p.Arg398Ter)	GBA	1	155236277	Gaucher disease, perinatal lethal;Gaucher's disease, type 1;not provided
NM_000157.3(GBA):c.476G>A (p.Arg159Gln)	GBA	1	155238629	Gaucher disease;Gaucher disease, perinatal lethal;Gaucher's disease, type 1
NM_000157.3(GBA):c.887G>A (p.Arg296Gln)	GBA	1	155237453	Gaucher disease;Gaucher disease, perinatal lethal;Gaucher's disease, type 1;not provided
NM_000157.3(GBA):c.1505G>A (p.Arg502His)	GBA	1	155235195	Gaucher disease;Gaucher's disease, type 1
NM_000157.3(GBA):c.1604G>A (p.Arg535His)	GBA	1	155235002	Gaucher disease;Gaucher's disease, type 1;not provided
NM_000157.3(GBA):c.475C>T (p.Arg159Trp)	GBA	1	155238630	Gaucher disease;Gaucher's disease, type 1;not provided
NM_000157.3(GBA):c.1246G>A (p.Gly416Ser)	GBA	1	155235823	Gaucher disease;Gaucher's disease, type 1;Subacute neuronopathic Gaucher's disease
NM_001005741.2(GBA):c.254G>A (p.Gly85Glu)	GBA	1	155239939	Gaucher's disease, type 1
NM_001005741.2(GBA):c.983C>T (p.Pro328Leu)	GBA	1	155237357	Gaucher's disease, type 1
NM_001005741.2(GBA):c.481C>T (p.Pro161Ser)	GBA	1	155238624	Gaucher's disease, type 1
NM_001005741.2(GBA):c.1549G>A (p.Gly517Ser)	GBA	1	155235057	Gaucher's disease, type 1
NM_001005741.2(GBA):c.1043C>T (p.Ala348Val)	GBA	1	155236426	Gaucher's disease, type 1
NM_001005741.2(GBA):c.259C>T (p.Arg87Trp)	GBA	1	155239934	Gaucher's disease, type 1
NM_001005741.2(GBA):c.1319C>T (p.Pro440Leu)	GBA	1	155235750	Gaucher's disease, type 1
NM_001005741.2(GBA):c.115+1G>A	GBA	1	155240629	Gaucher's disease, type 1;not provided
NM_001005741.2(GBA):c.508C>T (p.Arg170Cys)	GBA	1	155238597	Gaucher's disease, type 1;not provided
NM_001005741.2(GBA):c.625C>T (p.Arg209Cys)	GBA	1	155238270	Gaucher's disease, type 1;not provided
NM_001005741.2(GBA):c.721G>A (p.Gly241Arg)	GBA	1	155238174	Gaucher's disease, type 1;not provided

NM_022370.3(ROBO3):c.1082G>A (p.Gly361Glu)	ROBO3	11	124871062	Gaze palsy, familial horizontal, with progressive scoliosis
NM_022370.3(ROBO3):c.955G>A (p.Glu319Lys)	ROBO3	11	124870650	Gaze palsy, familial horizontal, with progressive scoliosis
NM_022370.3(ROBO3):c.733C>T (p.Arg245Trp)	ROBO3	11	124870035	Gaze palsy, familial horizontal, with progressive scoliosis
NM_022370.3(ROBO3):c.2317C>T (p.Gln773Ter)	ROBO3	11	124875581	Gaze palsy, familial horizontal, with progressive scoliosis
NM_001145320.1(ADAMTSL2):c.440C>T (p.Pro147Leu)	ADAMTSL2	9	133540625	Geleophysic dysplasia 1
NM_001145320.1(ADAMTSL2):c.338G>A (p.Arg113His)	ADAMTSL2	9	133539799	Geleophysic dysplasia 1
NM_001145320.1(ADAMTSL2):c.340G>A (p.Glu114Lys)	ADAMTSL2	9	133539801	Geleophysic dysplasia 1
NM_001145320.1(ADAMTSL2):c.2431G>A (p.Gly811Arg)	ADAMTSL2	9	133570346	Geleophysic dysplasia 1
NM_001145320.1(ADAMTSL2):c.2586G>A (p.Trp862Ter)	ADAMTSL2	9	133570501	Geleophysic dysplasia 1
NM_001145320.1(ADAMTSL2):c.215G>A (p.Arg72Gln)	ADAMTSL2	9	133537529	Geleophysic dysplasia 1
NM_001145320.1(ADAMTSL2):c.661C>T (p.Arg221Cys)	ADAMTSL2	9	133540980	Geleophysic dysplasia 1
NM_000138.4(FBN1):c.5284G>A (p.Gly1762Ser)	FBN1	15	48460258	Geleophysic dysplasia 2;Marfan syndrome;not provided
NM_001171.5(ABCC6):c.3736-1G>A	ABCC6	16	16157810	Generalized arterial calcification of infancy 2;Pseudoxanthoma elasticum
NM_001171.5(ABCC6):c.2294G>A (p.Arg765Gln)	ABCC6	16	16178919	Generalized arterial calcification of infancy 2;Pseudoxanthoma elasticum
NM_001171.5(ABCC6):c.3940C>T (p.Arg1314Trp)	ABCC6	16	16154974	Generalized arterial calcification of infancy 2;Pseudoxanthoma elasticum;not provided
NM_001171.5(ABCC6):c.1552C>T (p.Arg518Ter)	ABCC6	16	16190247	Generalized arterial calcification of infancy 2;Pseudoxanthoma elasticum;Pseudoxanthoma elasticum, forme fruste;not provided
NM_000094.3(COL7A1):c.6118G>A (p.Gly2040Ser)	COL7A1	3	48575401	Generalized dominant dystrophic epidermolysis bullosa
NM_000094.3(COL7A1):c.6017G>A (p.Gly2006Asp)	COL7A1	3	48575502	Generalized dominant dystrophic epidermolysis bullosa
NM_000094.3(COL7A1):c.6044G>A (p.Gly2015Glu)	COL7A1	3	48575475	Generalized dominant dystrophic epidermolysis bullosa
NM_000094.3(COL7A1):c.6227G>A (p.Gly2076Asp)	COL7A1	3	48575116	Generalized dominant dystrophic epidermolysis bullosa
NM_000094.3(COL7A1):c.6859G>A (p.Gly2287Arg)	COL7A1	3	48572712	Generalized dominant dystrophic epidermolysis bullosa;Nail disorder, nonsyndromic congenital, 8;Recessive dystrophic epidermolysis bullosa;not provided
NM_000094.3(COL7A1):c.6127G>A (p.Gly2043Arg)	COL7A1	3	48575392	Generalized dominant dystrophic epidermolysis bullosa;not provided
NM_000094.3(COL7A1):c.6100G>A (p.Gly2034Arg)	COL7A1	3	48575419	Generalized dominant dystrophic epidermolysis bullosa;not provided
NM_000094.3(COL7A1):c.6110G>A (p.Gly2037Glu)	COL7A1	3	48575409	Generalized dominant dystrophic epidermolysis bullosa;not provided
NM_198903.2(GABRG2):c.1312C>T (p.Gln438Ter)	GABRG2	5	162153132	Generalized epilepsy with febrile seizures plus 3
NM_000816.3(GABRG2):c.968G>A (p.Arg323Gln)	GABRG2	5	162149153	Generalized epilepsy with febrile seizures plus 3;not provided
NM_006920.4(SCN1A):c.2591C>T (p.Thr864Met)	SCN1A	2	166038098	Generalized epilepsy with febrile seizures plus, type 1;Generalized epilepsy with febrile seizures plus, type 2
NM_006920.4(SCN1A):c.2542C>T (p.Arg848Cys)	SCN1A	2	166039437	Generalized epilepsy with febrile seizures plus, type 1;not provided
NM_006920.4(SCN1A):c.1130G>A (p.Arg377Gln)	SCN1A	2	166047667	Generalized epilepsy with febrile seizures plus, type 1;Severe myoclonic epilepsy in infancy;not provided
NM_001165963.1(SCN1A):c.2214G>A (p.Trp738Ter)	SCN1A	2	166041432	Generalized epilepsy with febrile seizures plus, type 2;not provided
NM_006920.4(SCN1A):c.5093C>T (p.Thr1698Ile)	SCN1A	2	165992149	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.301C>T (p.Arg101Trp)	SCN1A	2	166058652	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.4186C>T (p.Arg1396Ter)	SCN1A	2	166002537	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.2584C>T (p.Arg862Ter)	SCN1A	2	166039428	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.4191G>A (p.Trp1397Ter)	SCN1A	2	166002532	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.4910G>A (p.Arg1637His)	SCN1A	2	165992332	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_001165963.1(SCN1A):c.664C>T (p.Arg222Ter)	SCN1A	2	166052882	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_006920.4(SCN1A):c.1177C>T (p.Arg393Cys)	SCN1A	2	166046970	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided

NM_006920.4(SCN1A):c.302G>A (p.Arg101Gln)	SCN1A	2	166058651	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_006920.4(SCN1A):c.5315C>T (p.Ala1772Val)	SCN1A	2	165991927	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_006920.4(SCN1A):c.2758C>T (p.Arg920Cys)	SCN1A	2	166037931	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_006920.4(SCN1A):c.2803C>T (p.Arg935Cys)	SCN1A	2	166037886	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_001165963.1(SCN1A):c.1837C>T (p.Arg613Ter)	SCN1A	2	166043875	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_001165963.1(SCN1A):c.2576G>A (p.Arg859His)	SCN1A	2	166039436	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_001165963.1(SCN1A):c.3733C>T (p.Arg1245Ter)	SCN1A	2	166012255	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_001165963.1(SCN1A):c.4906C>T (p.Arg1636Ter)	SCN1A	2	165992369	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_001165963.1(SCN1A):c.2134C>T (p.Arg712Ter)	SCN1A	2	166042334	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_001165963.1(SCN1A):c.602+1G>A	SCN1A	2	166054637	Generalized epilepsy with febrile seizures plus, type 2;Severe myoclonic epilepsy in infancy;not provided
NM_052874.4(STX1B):c.166C>T (p.Gln56Ter)	STX1B	16	31001133	Generalized epilepsy with febrile seizures plus, type 9
NM_002074.4(GNB1):c.976G>A (p.Ala326Thr)	GNB1	1	1787378	Generalized tonic-clonic seizures;Global developmental delay;Intellectual disability;Muscular hypotonia;Seizures
NM_000311.4(PRNP):c.305C>T (p.Pro102Leu)	PRNP	20	4699525	Genetic prion diseases;Gerstmann-Straussler-Scheinker syndrome
NM_000311.4(PRNP):c.350C>T (p.Ala117Val)	PRNP	20	4699570	Genetic prion diseases;Gerstmann-Straussler-Scheinker syndrome
NM_000311.4(PRNP):c.313C>T (p.Pro105Ser)	PRNP	20	4699533	Genetic prion diseases;Gerstmann-Straussler-Scheinker syndrome
NM_000311.4(PRNP):c.314C>T (p.Pro105Leu)	PRNP	20	4699534	Genetic prion diseases;Gerstmann-Straussler-Scheinker syndrome;Inborn genetic diseases
NM_000311.4(PRNP):c.628G>A (p.Val210Ile)	PRNP	20	4698848	Genetic prion diseases;Jakob-Creutzfeldt disease
NM_000311.4(PRNP):c.538G>A (p.Val180Ile)	PRNP	20	4699758	Genetic prion diseases;Jakob-Creutzfeldt disease
NM_000311.4(PRNP):c.623G>A (p.Arg208His)	PRNP	20	4698843	Genetic prion diseases;Jakob-Creutzfeldt disease
NM_152281.2(GORAB):c.859C>T (p.Arg287Ter)	GORAB	1	170552136	Geroderma osteodysplastica
NM_000311.4(PRNP):c.398C>T (p.Ala133Val)	PRNP	20	4699618	Gerstmann-Straussler-Scheinker syndrome
NM_000311.4(PRNP):c.679C>T (p.Gln227Ter)	PRNP	20	4699899	Gerstmann-Straussler-Scheinker syndrome
NM_001061.4(TBXAS1):c.1238G>A (p.Arg413Gln)	TBXAS1	7	140015731	Ghosal hematodiaphyseal syndrome
NM_022041.3(GAN):c.1456G>A (p.Glu486Lys)	GAN	16	81365432	Giant axonal neuropathy
NM_022041.3(GAN):c.1447C>T (p.Gln483Ter)	GAN	16	81365423	Giant axonal neuropathy
NM_022041.3(GAN):c.601C>T (p.Arg201Ter)	GAN	16	81354723	Giant axonal neuropathy
NM_022041.3(GAN):c.413G>A (p.Arg138His)	GAN	16	81354535	Giant axonal neuropathy
NM_022041.3(GAN):c.1429C>T (p.Arg477Ter)	GAN	16	81365405	Giant axonal neuropathy
NM_022041.3(GAN):c.505G>A (p.Glu169Lys)	GAN	16	81354627	Giant axonal neuropathy
NM_015726.3(DCAF8):c.949C>T (p.Arg317Cys)	DCAF8	1	160237145	Giant axonal neuropathy 2, autosomal dominant
NM_022041.3(GAN):c.851+1G>A	GAN	16	81357003	Giant axonal neuropathy;not provided
NM_001168272.1(ITPR1):c.4654C>T (p.Gln1552Ter)	ITPR1	3	4706208	Gillespie syndrome
NM_001168272.1(ITPR1):c.2137C>T (p.Arg713Ter)	ITPR1	3	4670904	Gillespie syndrome
NM_001168272.1(ITPR1):c.7615G>A (p.Gly2539Arg)	ITPR1	3	4814521	Gillespie syndrome

NM_000419.4(ITGA2B):c.1750C>T (p.Arg584Ter)	ITGA2B	17	44380004	Glanzmann thrombasthenia
NM_000419.4(ITGA2B):c.818G>A (p.Gly273Asp)	ITGA2B	17	44384567	Glanzmann thrombasthenia
NM_000419.4(ITGA2B):c.1544+1G>A	ITGA2B	17	44380385	Glanzmann thrombasthenia
NM_000419.4(ITGA2B):c.1073G>A (p.Arg358His)	ITGA2B	17	44383630	Glanzmann thrombasthenia
NM_000419.4(ITGA2B):c.1253G>A (p.Gly418Asp)	ITGA2B	17	44381019	Glanzmann thrombasthenia
NM_000419.4(ITGA2B):c.1063G>A (p.Glu355Lys)	ITGA2B	17	44383640	Glanzmann thrombasthenia
NM_000419.4(ITGA2B):c.2870C>T (p.Ser957Leu)	ITGA2B	17	44374732	Glanzmann thrombasthenia
NM_000212.2(ITGB3):c.719G>A (p.Arg240Gln)	ITGB3	17	47286364	Glanzmann thrombasthenia
NM_000212.2(ITGB3):c.718C>T (p.Arg240Trp)	ITGB3	17	47286363	Glanzmann thrombasthenia
NM_000212.2(ITGB3):c.1199G>A (p.Cys400Tyr)	ITGB3	17	47291027	Glanzmann thrombasthenia
NM_000212.2(ITGB3):c.2248C>T (p.Arg750Ter)	ITGB3	17	47307584	Glanzmann thrombasthenia
NM_000212.2(ITGB3):c.740G>A (p.Gly247Asp)	ITGB3	17	47286385	Glanzmann thrombasthenia
NM_000261.1(MYOC):c.136C>T (p.Arg46Ter)	MYOC	1	171652476	Glaucoma 1, open angle, a, autosomal recessive
NM_001008211.1(OPTN):c.148G>A (p.Glu50Lys)	OPTN	10	13109270	Glaucoma 1, open angle, e
NM_080871.3(ASB10):c.765C>T (p.Thr255=)	ASB10	7	151181233	Glaucoma 1, open angle, f
NM_001142459.1(ASB10):c.215G>A (p.Arg72His)	ASB10	7	151186916	Glaucoma 1, open angle, f
NM_006179.4(NTF4):c.616C>T (p.Arg206Trp)	NTF4	19	49061382	Glaucoma 1, open angle, O
NM_006179.4(NTF4):c.617G>A (p.Arg206Gln)	NTF4	19	49061381	Glaucoma 1, open angle, O
NM_000428.2(LTBP2):c.331C>T (p.Gln111Ter)	LTBP2	14	74611614	Glaucoma 3, primary congenital, d
NM_000428.2(LTBP2):c.895C>T (p.Arg299Ter)	LTBP2	14	74555629	Glaucoma 3, primary congenital, d;Microspherophakia
NM_000104.3(CYP1B1):c.1120G>A (p.Asp374Asn)	CYP1B1	2	38071234	Glaucoma, congenital
NM_000104.3(CYP1B1):c.1159G>A (p.Glu387Lys)	CYP1B1	2	38071195	Glaucoma, congenital
NM_000104.3(CYP1B1):c.182G>A (p.Gly61Glu)	CYP1B1	2	38075207	Glaucoma, congenital;not provided
NM_000104.3(CYP1B1):c.1405C>T (p.Arg469Trp)	CYP1B1	2	38070949	Glaucoma, congenital;not provided
NM_030751.5(ZEB1):c.973C>T (p.Arg325Ter)	ZEB1	10	31520308	Glaucoma;Polymorpheus posterior corneal dystrophy;Visual loss
NM_000404.3(GLB1):c.602G>A (p.Arg201His)	GLB1	3	33058220	GLB1-Related Disorders;GM1 gangliosidosis type 2;Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis, MPS-IV-B;not provided
NM_001005862.2(ERBB2):c.2650G>A (p.Glu884Lys)	ERBB2	17	39725721	Glioma susceptibility 1
NM_000314.6(PTEN):c.701G>A (p.Arg234Gln)	PTEN	10	87957919	Glioma susceptibility 2;Meningioma
NM_012160.4(FBXL4):c.1444C>T (p.Arg482Trp)	FBXL4	6	98875673	Global developmental delay;Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type);Mitochondrial encephalomyopathy;not provided
NM_002074.4(GNB1):c.229G>A (p.Gly77Ser)	GNB1	1	1806513	Global developmental delay;Muscular hypotonia
NM_031955.5(SPATA16):c.848G>A (p.Arg283Gln)	SPATA16	3	173019486	Globozoospermia
NM_206898.1(MRAP):c.3G>A (p.Met1Ile)	MRAP	21	32298974	Glucocorticoid deficiency 2
NM_012343.3(NNT):c.1598C>T (p.Ala533Val)	NNT	5	43649300	Glucocorticoid deficiency 4
NM_012343.3(NNT):c.385C>T (p.Arg129Ter)	NNT	5	43615851	Glucocorticoid deficiency 4
NM_012343.3(NNT):c.211C>T (p.Arg71Ter)	NNT	5	43612967	Glucocorticoid deficiency 4
NM_015665.5(AAAS):c.934C>T (p.Arg312Ter)	AAAS	12	53309158	Glucocorticoid deficiency with achalasia
NM_015665.5(AAAS):c.1432C>T (p.Arg478Ter)	AAAS	12	53307698	Glucocorticoid deficiency with achalasia;not provided
NM_001018077.1(NR3C1):c.1430G>A (p.Arg477His)	NR3C1	5	143310135	Glucocorticoid resistance, generalized
NM_001018077.1(NR3C1):c.2035G>A (p.Gly679Ser)	NR3C1	5	143282714	Glucocorticoid resistance, generalized
NM_001164277.1(SLC37A4):c.1243C>T (p.Arg415Ter)	SLC37A4	11	119024957	Glucose-6-phosphate transport defect
NM_001164277.1(SLC37A4):c.83G>A (p.Arg28His)	SLC37A4	11	119029287	Glucose-6-phosphate transport defect;not provided
NM_001164277.1(SLC37A4):c.1016G>A (p.Gly339Asp)	SLC37A4	11	119025298	Glucose-6-phosphate transport defect;not provided
NM_001164277.1(SLC37A4):c.742C>T (p.Gln248Ter)	SLC37A4	11	119026979	Glucose-6-phosphate transport defect;not provided

NM_001164277.1(SLC37A4):c.287G>A (p.Trp96Ter)	SLC37A4	11	119028288	Glucose-6-phosphate transport defect;Phosphate transport defect
NM_006516.2(SLC2A1):c.272G>A (p.Gly91Asp)	SLC2A1	1	42931049	GLUT1 deficiency syndrome 1
NM_006516.2(SLC2A1):c.847C>T (p.Gln283Ter)	SLC2A1	1	42929613	GLUT1 deficiency syndrome 1
NM_006516.2(SLC2A1):c.1402C>T (p.Arg468Trp)	SLC2A1	1	42927118	GLUT1 deficiency syndrome 1, autosomal recessive
NM_006516.2(SLC2A1):c.988C>T (p.Arg330Ter)	SLC2A1	1	42929018	GLUT1 deficiency syndrome 1, autosomal recessive;Microcephaly;Seizure Disorders;intellectual deficiency;not provided
NM_006516.2(SLC2A1):c.1198C>T (p.Arg400Cys)	SLC2A1	1	42927685	GLUT1 deficiency syndrome 1, autosomal recessive;not provided
NM_006516.2(SLC2A1):c.971C>T (p.Ser324Leu)	SLC2A1	1	42929211	GLUT1 deficiency syndrome 1, autosomal recessive;not provided
NM_006516.2(SLC2A1):c.823G>A (p.Ala275Thr)	SLC2A1	1	42929637	GLUT1 deficiency syndrome 1;GLUT1 deficiency syndrome 2;not provided
NM_006516.2(SLC2A1):c.724C>T (p.Gln242Ter)	SLC2A1	1	42929736	GLUT1 deficiency syndrome 1;Intellectual disability
NM_006516.2(SLC2A1):c.748C>T (p.Gln250Ter)	SLC2A1	1	42929712	GLUT1 deficiency syndrome 1;not provided
NM_006516.2(SLC2A1):c.884C>T (p.Thr295Met)	SLC2A1	1	42929298	GLUT1 deficiency syndrome 1;not provided
NM_006516.2(SLC2A1):c.277C>T (p.Arg93Trp)	SLC2A1	1	42930865	GLUT1 deficiency syndrome 2;not provided
NM_006516.2(SLC2A1):c.274C>T (p.Arg92Trp)	SLC2A1	1	42931047	GLUT1 deficiency syndrome 2;not provided
NM_006657.2(FTCD):c.403C>T (p.Arg135Cys)	FTCD	21	46151945	GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY
NM_002065.6(GLUL):c.970C>T (p.Arg324Cys)	GLUL	1	182384557	Glutamine deficiency, congenital
NM_002065.6(GLUL):c.1021C>T (p.Arg341Cys)	GLUL	1	182384506	Glutamine deficiency, congenital
NM_000126.3(ETFA):c.346G>A (p.Gly116Arg)	ETFA	15	76292436	Glutaric acidemia IIA
NM_000126.3(ETFA):c.797C>T (p.Thr266Met)	ETFA	15	76274431	Glutaric acidemia IIA;Glutaric aciduria, type 2;not provided
NM_001985.2(ETFB):c.491G>A (p.Arg164Gln)	ETFB	19	51347006	Glutaric acidemia IIB
NM_001985.2(ETFB):c.382G>A (p.Asp128Asn)	ETFB	19	51350385	Glutaric acidemia IIB
NM_004453.3(ETFDH):c.524G>A (p.Arg175His)	ETFDH	4	158685137	Glutaric acidemia IIC
NM_004453.3(ETFDH):c.1448C>T (p.Pro483Leu)	ETFDH	4	158706351	Glutaric acidemia iic, late-onset
NM_004453.3(ETFDH):c.250G>A (p.Ala84Thr)	ETFDH	4	158682269	Glutaric acidemia IIC;not provided
NM_000159.3(GCDH):c.1262C>T (p.Ala421Val)	GCDH	19	12899486	Glutaric acidemia;Glutaric aciduria, type 1;not provided
NM_000159.3(GCDH):c.1204C>T (p.Arg402Trp)	GCDH	19	12897824	Glutaric acidemia;Glutaric aciduria, type 1;not provided
NM_000159.3(GCDH):c.1247C>T (p.Thr416Ile)	GCDH	19	12899471	Glutaric aciduria, type 1
NM_000159.3(GCDH):c.482G>A (p.Arg161Gln)	GCDH	19	12893630	Glutaric aciduria, type 1
NM_000159.3(GCDH):c.675G>A (p.Trp225Ter)	GCDH	19	12896244	Glutaric aciduria, type 1
NM_000159.3(GCDH):c.382C>T (p.Arg128Ter)	GCDH	19	12893530	Glutaric aciduria, type 1
NM_000159.3(GCDH):c.877G>A (p.Ala293Thr)	GCDH	19	12896934	Glutaric aciduria, type 1;not provided
NM_000159.3(GCDH):c.1093G>A (p.Glu365Lys)	GCDH	19	12897713	Glutaric aciduria, type 1;not provided
NM_000159.3(GCDH):c.1198G>A (p.Val400Met)	GCDH	19	12897818	Glutaric aciduria, type 1;not provided
NM_000159.3(GCDH):c.636-1G>A	GCDH	19	12896204	Glutaric aciduria, type 1;not provided
NM_000159.3(GCDH):c.1240G>A (p.Glu414Lys)	GCDH	19	12897860	Glutaric aciduria, type 1;not provided
NM_000159.3(GCDH):c.262C>T (p.Arg88Cys)	GCDH	19	12891965	Glutaric aciduria, type 1;not provided
NM_000159.3(GCDH):c.416C>T (p.Ser139Leu)	GCDH	19	12893564	Glutaric aciduria, type 1;not provided
NM_025207.4(FLAD1):c.1588C>T (p.Arg530Cys)	FLAD1	1	154992746	Glutaric aciduria, type 2;Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency
NM_004453.3(ETFDH):c.1367C>T (p.Pro456Leu)	ETFDH	4	158706270	Glutaric aciduria, type 2;not provided
NM_001193311.1(SUGCT):c.535C>T (p.Arg179Ter)	SUGCT	7	40237664	Glutaryl-CoA oxidase deficiency
NM_001193311.1(SUGCT):c.322C>T (p.Arg108Ter)	SUGCT	7	40188569	Glutaryl-CoA oxidase deficiency
NM_024728.2(SUGCT):c.895C>T (p.Arg299Trp)	SUGCT	7	40459197	Glutaryl-CoA oxidase deficiency;not provided
NM_000178.3(GSS):c.491G>A (p.Arg164Gln)	GSS	20	34942488	Glutathione synthetase deficiency;Gluthathione synthetase deficiency
NM_000178.3(GSS):c.799C>T (p.Arg267Trp)	GSS	20	34935611	Gluthathione synthetase deficiency

NM_000178.3(GSS):c.847C>T (p.Arg283Cys)	GSS	20	34932121	Gluthathione synthetase deficiency
NM_000178.3(GSS):c.373C>T (p.Arg125Cys)	GSS	20	34942606	Gluthathione synthetase deficiency
NM_000178.3(GSS):c.941C>T (p.Pro314Leu)	GSS	20	34932027	Gluthathione synthetase deficiency
NM_001024845.2(SLC6A9):c.1498C>T (p.Gln500Ter)	SLC6A9	1	44000805	GLYCINE ENCEPHALOPATHY WITH NORMAL SERUM GLYCINE
NM_017431.2(PRKAG3):c.673C>T (p.Arg225Trp)	PRKAG3	2	218828561	Glycogen content in skeletal muscle, increased
NM_000293.2(PHKB):c.1969C>T (p.Gln657Ter)	PHKB	16	47650919	Glycogen phosphorylase kinase deficiency;Glycogen storage disease IXb
NM_002103.4(GYS1):c.1384C>T (p.Arg462Ter)	GYS1	19	48974658	Glycogen storage disease 0, muscle
NM_000152.4(GAA):c.1634C>T (p.Pro545Leu)	GAA	17	80111023	Glycogen storage disease II, adult form
NM_000152.4(GAA):c.710C>T (p.Ala237Val)	GAA	17	80107574	Glycogen storage disease II, adult form
NM_000152.4(GAA):c.877G>A (p.Gly293Arg)	GAA	17	80107818	Glycogen storage disease II, adult form
NM_000152.4(GAA):c.1927G>A (p.Gly643Arg)	GAA	17	80112914	Glycogen storage disease II, adult form;Glycogen storage disease, type II
NM_000152.4(GAA):c.2173C>T (p.Arg725Trp)	GAA	17	80113350	Glycogen storage disease II, adult form;Glycogen storage disease, type II
NM_000642.2(AGL):c.1222C>T (p.Arg408Ter)	AGL	1	99875394	Glycogen storage disease IIIa;Glycogen storage disease type III
NM_000642.2(AGL):c.3980G>A (p.Trp1327Ter)	AGL	1	99913557	Glycogen storage disease IIIa;Glycogen storage disease type III;not provided
NM_000642.2(AGL):c.16C>T (p.Gln6Ter)	AGL	1	99851058	Glycogen storage disease IIIb;Glycogen storage disease type III
NM_000642.2(AGL):c.2039G>A (p.Trp680Ter)	AGL	1	99881329	Glycogen storage disease IIIb;Glycogen storage disease type III
NM_000158.3(GBE1):c.783-1G>A	GBE1	3	81642991	Glycogen storage disease IV, congenital neuromuscular
NM_000158.3(GBE1):c.784C>T (p.Arg262Cys)	GBE1	3	81642989	Glycogen storage disease IV, congenital neuromuscular
NM_000158.3(GBE1):c.1643G>A (p.Trp548Ter)	GBE1	3	81537071	Glycogen storage disease IV, congenital neuromuscular;Glycogen storage disease, type IV
NM_000292.2(PHKA2):c.3341C>T (p.Thr1114Ile)	PHKA2	X	18894400	Glycogen storage disease IXa2
NM_000292.2(PHKA2):c.557G>A (p.Arg186His)	PHKA2	X	18945139	Glycogen storage disease IXa2
NM_000292.2(PHKA2):c.394C>T (p.His132Tyr)	PHKA2	X	18951164	Glycogen storage disease IXa2
NM_000292.2(PHKA2):c.556C>T (p.Arg186Cys)	PHKA2	X	18945140	Glycogen storage disease IXa2
NM_000293.2(PHKB):c.1546C>T (p.Gln516Ter)	PHKB	16	47641630	Glycogen storage disease IXb
NM_000294.2(PHKG2):c.566G>A (p.Gly189Glu)	PHKG2	16	30756191	Glycogen storage disease IXc
NM_000294.2(PHKG2):c.130C>T (p.Arg44Ter)	PHKG2	16	30751140	Glycogen storage disease IXc
NM_000294.2(PHKG2):c.433C>T (p.His145Tyr)	PHKG2	16	30753434	Glycogen storage disease IXc
NM_002637.3(PHKA1):c.667G>A (p.Gly223Arg)	PHKA1	X	72667425	Glycogen storage disease IXd
NM_016203.3(PRKAG2):c.1592G>A (p.Arg531Gln)	PRKAG2	7	151560610	Glycogen storage disease of heart, lethal congenital;Primary familial hypertrophic cardiomyopathy;not provided
NM_001976.4(ENO3):c.467G>A (p.Gly156Asp)	ENO3	17	4955097	Glycogen storage disease type 13
NM_001976.4(ENO3):c.1121G>A (p.Gly374Glu)	ENO3	17	4956626	Glycogen storage disease type 13
NM_000151.3(G6PC):c.328G>A (p.Glu110Lys)	G6PC	17	42904028	Glycogen storage disease type 1A
NM_000151.3(G6PC):c.370G>A (p.Ala124Thr)	G6PC	17	42907552	Glycogen storage disease type 1A
NM_000151.3(G6PC):c.551G>A (p.Gly184Glu)	G6PC	17	42909407	Glycogen storage disease type 1A
NM_000151.3(G6PC):c.248G>A (p.Arg83His)	G6PC	17	42903948	Glycogen storage disease type 1A
NM_000151.3(G6PC):c.724C>T (p.Gln242Ter)	G6PC	17	42911076	Glycogen storage disease type 1A
NM_000151.3(G6PC):c.883C>T (p.Arg295Cys)	G6PC	17	42911235	Glycogen storage disease type 1A;Glycogen storage disease, type I
NM_000151.3(G6PC):c.247C>T (p.Arg83Cys)	G6PC	17	42903947	Glycogen storage disease type 1A;Glycogen storage disease, type I;not provided
NM_000151.3(G6PC):c.1039C>T (p.Gln347Ter)	G6PC	17	42911391	Glycogen storage disease type 1A;not provided
NM_000152.4(GAA):c.1561G>A (p.Glu521Lys)	GAA	17	80110950	Glycogen storage disease type II, infantile;Glycogen storage disease, type II
NM_000642.2(AGL):c.2590C>T (p.Arg864Ter)	AGL	1	99884612	Glycogen storage disease type III
NM_000642.2(AGL):c.3682C>T (p.Arg1228Ter)	AGL	1	99902776	Glycogen storage disease type III
NM_000642.2(AGL):c.118C>T (p.Gln40Ter)	AGL	1	99861538	Glycogen storage disease type III
NM_000642.2(AGL):c.256C>T (p.Gln86Ter)	AGL	1	99861676	Glycogen storage disease type III

NM_000642.2(AGL):c.2681+1G>A	AGL	1	99884704	Glycogen storage disease type III
NM_000642.2(AGL):c.2158-1G>A	AGL	1	99881540	Glycogen storage disease type III
NM_000028.2(AGL):c.3836+1G>A	AGL	1	99910848	Glycogen storage disease type III
NM_000292.2(PHKA2):c.3025C>T (p.Gln1009Ter)	PHKA2	X	18901487	Glycogen storage disease type IXa1
NM_000292.2(PHKA2):c.2296C>T (p.Gln766Ter)	PHKA2	X	18908865	Glycogen storage disease type IXa1
NM_000292.2(PHKA2):c.3614C>T (p.Pro1205Leu)	PHKA2	X	18893579	Glycogen storage disease type IXa1
NM_000292.2(PHKA2):c.883C>T (p.Arg295Cys)	PHKA2	X	18940030	Glycogen storage disease type IXa1
NM_000290.3(PGAM2):c.233G>A (p.Trp78Ter)	PGAM2	7	44065297	Glycogen storage disease type X
NM_000290.3(PGAM2):c.290G>A (p.Gly97Asp)	PGAM2	7	44065240	Glycogen storage disease type X
NM_004130.3(GYG1):c.248C>T (p.Thr83Met)	GYG1	3	148996406	Glycogen storage disease XV
NM_000152.4(GAA):c.1004G>A (p.Gly335Glu)	GAA	17	80108338	Glycogen storage disease, type II
NM_000152.4(GAA):c.1437G>A (p.Lys479=)	GAA	17	80110055	Glycogen storage disease, type II
NM_000152.4(GAA):c.1548G>A (p.Trp516Ter)	GAA	17	80110837	Glycogen storage disease, type II
NM_000152.4(GAA):c.2014C>T (p.Arg672Trp)	GAA	17	80113001	Glycogen storage disease, type II
NM_000152.4(GAA):c.1802C>T (p.Ser601Leu)	GAA	17	80112625	Glycogen storage disease, type II
NM_000152.4(GAA):c.546G>A (p.Thr182=)	GAA	17	80105132	Glycogen storage disease, type II
NM_000152.4(GAA):c.1754+1G>A	GAA	17	80112101	Glycogen storage disease, type II
NM_000152.4(GAA):c.1082C>T (p.Pro361Leu)	GAA	17	80108495	Glycogen storage disease, type II
NM_000152.4(GAA):c.2560C>T (p.Arg854Ter)	GAA	17	80118271	Glycogen storage disease, type II;not provided
NM_000152.4(GAA):c.1465G>A (p.Asp489Asn)	GAA	17	80110754	Glycogen storage disease, type II;not provided
NM_000152.4(GAA):c.2512C>T (p.Gln838Ter)	GAA	17	80118223	Glycogen storage disease, type II;not provided
NM_000152.4(GAA):c.655G>A (p.Gly219Arg)	GAA	17	80105857	Glycogen storage disease, type II;not provided
NM_000152.4(GAA):c.1933G>A (p.Asp645Asn)	GAA	17	80112920	Glycogen storage disease, type II;not provided
NM_000152.4(GAA):c.1979G>A (p.Arg660His)	GAA	17	80112966	Glycogen storage disease, type II;not provided
NM_000158.3(GBE1):c.1570C>T (p.Arg524Ter)	GBE1	3	81577973	Glycogen storage disease, type IV
NM_000158.3(GBE1):c.1571G>A (p.Arg524Gln)	GBE1	3	81577972	Glycogen storage disease, type IV
NM_000158.3(GBE1):c.143+1G>A	GBE1	3	81761374	Glycogen storage disease, type IV
NM_000158.3(GBE1):c.1543C>T (p.Arg515Cys)	GBE1	3	81578000	Glycogen storage disease, type IV;inborn genetic diseases
NM_005609.3(PYGM):c.1963G>A (p.Glu655Lys)	PYGM	11	64751331	Glycogen storage disease, type V
NM_005609.3(PYGM):c.2056G>A (p.Gly686Arg)	PYGM	11	64750497	Glycogen storage disease, type V
NM_005609.3(PYGM):c.1726C>T (p.Arg576Ter)	PYGM	11	64751966	Glycogen storage disease, type V
NM_005609.3(PYGM):c.1827G>A (p.Lys609=)	PYGM	11	64751597	Glycogen storage disease, type V
NM_005609.3(PYGM):c.148C>T (p.Arg50Ter)	PYGM	11	64759751	Glycogen storage disease, type V;not provided
NM_005609.3(PYGM):c.613G>A (p.Gly205Ser)	PYGM	11	64757826	Glycogen storage disease, type V;not provided
NM_005609.3(PYGM):c.1366G>A (p.Val456Met)	PYGM	11	64753556	Glycogen storage disease, type V;not provided
NM_005609.3(PYGM):c.1768+1G>A	PYGM	11	64751923	Glycogen storage disease, type V;not provided
NM_002863.4(PYGL):c.1768+1G>A	PYGL	14	50912155	Glycogen storage disease, type VI
NM_002863.4(PYGL):c.1620+1G>A	PYGL	14	50913028	Glycogen storage disease, type VI
NM_002863.4(PYGL):c.1195C>T (p.Arg399Ter)	PYGL	14	50915869	Glycogen storage disease, type VI
NM_002863.4(PYGL):c.1366G>A (p.Val456Met)	PYGL	14	50915373	Glycogen storage disease, type VI
NM_002863.4(PYGL):c.1471C>T (p.Arg491Cys)	PYGL	14	50914748	Glycogen storage disease, type VI
NM_002863.4(PYGL):c.2017G>A (p.Glu673Lys)	PYGL	14	50910055	Glycogen storage disease, type VI
NM_002863.4(PYGL):c.2024C>T (p.Ser675Leu)	PYGL	14	50910048	Glycogen storage disease, type VI
NM_002863.4(PYGL):c.280C>T (p.Arg94Ter)	PYGL	14	50937801	Glycogen storage disease, type VI
NM_002863.4(PYGL):c.698G>A (p.Gly233Asp)	PYGL	14	50921030	Glycogen storage disease, type VI

NM_001166686.1(PFKM):c.496C>T (p.Arg166Ter)	PFKM	12	48132913	Glycogen storage disease, type VII
NM_001166686.1(PFKM):c.450+1G>A	PFKM	12	48131394	Glycogen storage disease, type VII
NM_000404.3(GLB1):c.202C>T (p.Arg68Trp)	GLB1	3	33072587	GM1 gangliosidosis type 2
NM_000404.3(GLB1):c.1768C>T (p.Arg590Cys)	GLB1	3	32997311	GM1 gangliosidosis type 2;Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis, MPS-IV-B
NM_000404.3(GLB1):c.1445G>A (p.Arg482His)	GLB1	3	33016743	GM1 gangliosidosis type 2;Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis, MPS-IV-B;not provided
NM_000404.3(GLB1):c.622C>T (p.Arg208Cys)	GLB1	3	33058200	GM1 gangliosidosis type 2;Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis, MPS-IV-B;not provided
NM_000404.3(GLB1):c.1769G>A (p.Arg590His)	GLB1	3	32997310	GM1 gangliosidosis type 2;Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis, MPS-IV-B;not provided
NM_000404.3(GLB1):c.397-1G>A	GLB1	3	33068291	GM1 gangliosidosis type 2;Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis, MPS-IV-B;not provided
NM_000404.3(GLB1):c.442C>T (p.Arg148Cys)	GLB1	3	33068245	GM1 gangliosidosis type 2;Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis, MPS-IV-B;not provided
NM_000404.3(GLB1):c.176G>A (p.Arg59His)	GLB1	3	33072613	GM1 gangliosidosis type 2;GM1-gangliosidosis, type I, with cardiac involvement;Gangliosidosis GM1 type 3;Infantile GM1 gangliosidosis;Mucopolysaccharidosis, MPS-IV-B;not provided
NM_000404.3(GLB1):c.1051C>T (p.Arg351Ter)	GLB1	3	33046137	GM1-gangliosidosis, type I, with cardiac involvement
NM_000520.5(HEXA):c.805G>A (p.Gly269Ser)	HEXA	15	72350518	Gm2-gangliosidosis, adult;Tay-Sachs disease;not provided
NM_000520.5(HEXA):c.1495C>T (p.Arg499Cys)	HEXA	15	72345477	Gm2-gangliosidosis, adult-onset;Tay-Sachs disease
NM_000520.5(HEXA):c.1510C>T (p.Arg504Cys)	HEXA	15	72345462	Gm2-gangliosidosis, chronic;Tay-Sachs disease
NM_000520.5(HEXA):c.806-7G>A	HEXA	15	72349266	Gm2-gangliosidosis, chronic;Tay-Sachs disease
NM_000520.5(HEXA):c.1496G>A (p.Arg499His)	HEXA	15	72345476	Gm2-gangliosidosis, juvenile;Inborn genetic diseases;Tay-Sachs disease
NM_000520.5(HEXA):c.1511G>A (p.Arg504His)	HEXA	15	72345461	Gm2-gangliosidosis, juvenile;Tay-Sachs disease
NM_000520.5(HEXA):c.987G>A (p.Trp329Ter)	HEXA	15	72348134	Gm2-gangliosidosis, variant b1
NM_213599.2(ANO5):c.1541C>T (p.Thr514Ile)	ANO5	11	22259652	Gnathodiaphyseal dysplasia
NM_024312.4(GNPTAB):c.1123C>T (p.Arg375Ter)	GNPTAB	12	101770182	GNPTAB-Related Disorders;I cell disease;not provided
NM_177438.2(DICER1):c.2516C>T (p.Ser839Phe)	DICER1	14	95108014	Goiter, multinodular 1, with or without sertoli-leydig cell tumors
NM_000414.3(HSD17B4):c.101C>T (p.Ala34Val)	HSD17B4	5	119456357	Gonadal dysgenesis with auditory dysfunction, autosomal recessive inheritance
NM_000414.3(HSD17B4):c.1538C>T (p.Pro513Leu)	HSD17B4	5	119525250	Gonadal dysgenesis with auditory dysfunction, autosomal recessive inheritance
NM_000233.3(LHCGR):c.1713G>A (p.Met571Ile)	LHCGR	2	48688084	Gonadotropin-independent familial sexual precocity
NM_000233.3(LHCGR):c.1730C>T (p.Thr577Ile)	LHCGR	2	48688067	Gonadotropin-independent familial sexual precocity
NM_000233.3(LHCGR):c.1715C>T (p.Ala572Val)	LHCGR	2	48688082	Gonadotropin-independent familial sexual precocity
NM_000233.3(LHCGR):c.1118C>T (p.Ala373Val)	LHCGR	2	48688679	Gonadotropin-independent familial sexual precocity
NM_000233.3(LHCGR):c.1703C>T (p.Ala568Val)	LHCGR	2	48688094	Gonadotropin-independent familial sexual precocity
NM_207111.3(RNF216):c.2251C>T (p.Arg751Cys)	RNF216	7	5641285	Gordon Holmes syndrome
NM_207111.3(RNF216):c.1367G>A (p.Gly456Glu)	RNF216	7	5729454	Gordon Holmes syndrome
NM_207111.3(RNF216):c.904C>T (p.Gln302Ter)	RNF216	7	5741113	Gordon Holmes syndrome
NM_022068.3(PIEZO2):c.8057G>A (p.Arg2686His)	PIEZO2	18	10671729	Gordon's syndrome;Oculomelic amyoplasia
NM_001166292.1(PTCH2):c.2156G>A (p.Arg719Gln)	PTCH2	1	44827617	Gorlin syndrome
NM_000264.3(PTCH1):c.1081C>T (p.Gln361Ter)	PTCH1	9	95479134	Gorlin syndrome
NM_000264.3(PTCH1):c.945+1G>A	PTCH1	9	95480389	Gorlin syndrome
NM_000264.3(PTCH1):c.3152G>A (p.Trp1051Ter)	PTCH1	9	95458029	Gorlin syndrome
NM_000264.3(PTCH1):c.2308C>T (p.Arg770Ter)	PTCH1	9	95467368	Gorlin syndrome
NM_000264.3(PTCH1):c.1882C>T (p.Gln628Ter)	PTCH1	9	95469119	Gorlin syndrome

NM_000264.3(PTCH1):c.708G>A (p.Trp236Ter)	PTCH1	9	95481987	Gorlin syndrome
NM_016169.3(SUFU):c.436C>T (p.Arg146Ter)	SUFU	10	102550088	Gorlin syndrome;Medulloblastoma
NM_016169.3(SUFU):c.1022+1G>A	SUFU	10	102599545	Gorlin syndrome;Medulloblastoma, desmoplastic
NM_000101.3(CYBA):c.269G>A (p.Arg90Gln)	CYBA	16	88646773	Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative
NM_000101.3(CYBA):c.7C>T (p.Gln3Ter)	CYBA	16	88651007	Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative
NM_000101.3(CYBA):c.70G>A (p.Gly24Arg)	CYBA	16	88648103	Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative
NM_000101.3(CYBA):c.373G>A (p.Ala125Thr)	CYBA	16	88643568	Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative
NM_000397.3(CYBB):c.466G>A (p.Ala156Thr)	CYBB	X	37793793	Granulomatous disease, chronic, X-linked, variant;not provided
NM_000397.3(CYBB):c.301C>T (p.His101Tyr)	CYBB	X	37792023	Granulomatous disease, chronic, X-linked, variant;not provided
NM_015175.2(NBEAL2):c.2701C>T (p.Arg901Ter)	NBEAL2	3	46997310	Gray platelet syndrome
NM_015175.2(NBEAL2):c.6299C>T (p.Pro2100Leu)	NBEAL2	3	47004976	Gray platelet syndrome
NM_015175.2(NBEAL2):c.1823G>A (p.Trp608Ter)	NBEAL2	3	46995558	Gray platelet syndrome
NM_000557.4(GDF5):c.1199G>A (p.Cys400Tyr)	GDF5	20	35434216	Grebe syndrome
NM_194442.2(LBR):c.1748G>A (p.Arg583Gln)	LBR	1	225403403	Greenberg dysplasia
NM_000168.5(GLI3):c.1873C>T (p.Arg625Trp)	GLI3	7	41972567	Greig cephalopolysyndactyly syndrome
NM_000168.5(GLI3):c.2374C>T (p.Arg792Ter)	GLI3	7	41967653	Greig cephalopolysyndactyly syndrome
NM_000168.5(GLI3):c.1486C>T (p.Gln496Ter)	GLI3	7	42023479	Greig cephalopolysyndactyly syndrome
NM_000168.5(GLI3):c.2119C>T (p.Pro707Ser)	GLI3	7	41967908	Greig cephalopolysyndactyly syndrome
NM_000168.5(GLI3):c.868C>T (p.Arg290Ter)	GLI3	7	42040198	Greig cephalopolysyndactyly syndrome;Preaxial polydactyly 4
NM_183235.2(RAB27A):c.352C>T (p.Gln118Ter)	RAB27A	15	55224004	Griscelli syndrome type 2
NM_024101.6(MLPH):c.103C>T (p.Arg35Trp)	MLPH	2	237493529	Griscelli syndrome type 3
NM_000358.2(TGFBI):c.1663C>T (p.Arg555Trp)	TGFBI	5	136056780	Groenouw corneal dystrophy type I
NM_004247.3(EFTUD2):c.784C>T (p.Arg262Trp)	EFTUD2	17	44876019	Growth and mental retardation, mandibulofacial dysostosis, microcephaly, and cleft palate
NM_004247.3(EFTUD2):c.2770C>T (p.Gln924Ter)	EFTUD2	17	44851763	Growth and mental retardation, mandibulofacial dysostosis, microcephaly, and cleft palate
NM_018129.3(PNPO):c.674G>A (p.Arg225His)	PNPO	17	47946670	Growth delay;Intrauterine growth retardation;Pyridoxal 5'-phosphate-dependent epilepsy;Seizures
NM_003865.2(HESX1):c.445G>A (p.Glu149Lys)	HESX1	3	57198405	Growth hormone deficiency with pituitary anomalies
NM_012448.3(STAT5B):c.454C>T (p.Arg152Ter)	STAT5B	17	42223478	Growth hormone insensitivity with immunodeficiency
NM_001080432.2(FTO):c.947G>A (p.Arg316Gln)	FTO	16	53873837	Growth retardation, developmental delay, coarse facies, and early death
NM_001080432.2(FTO):c.956C>T (p.Ser319Phe)	FTO	16	53873846	Growth retardation, developmental delay, coarse facies, and early death
NM_013417.3(IARS):c.1252C>T (p.Arg418Ter)	IARS	9	92269937	GROWTH RETARDATION, INTELLECTUAL DEVELOPMENTAL DISORDER, HYPOTONIA, AND HEPATOPATHY
NM_013417.3(IARS):c.1310C>T (p.Pro437Leu)	IARS	9	92268295	GROWTH RETARDATION, INTELLECTUAL DEVELOPMENTAL DISORDER, HYPOTONIA, AND HEPATOPATHY
NM_013417.3(IARS):c.760C>T (p.Arg254Ter)	IARS	9	92278272	GROWTH RETARDATION, INTELLECTUAL DEVELOPMENTAL DISORDER, HYPOTONIA, AND HEPATOPATHY
NM_000161.2(GCH1):c.633G>A (p.Met211Ile)	GCH1	14	54844137	GTP cyclohydrolase I deficiency
NM_000161.2(GCH1):c.551G>A (p.Arg184His)	GCH1	14	54845843	GTP cyclohydrolase I deficiency
NM_000274.3(OAT):c.677C>T (p.Ala226Val)	OAT	10	124403892	Gyrate atrophy of choroid and retina with pyridoxine-responsive ornithinemia
NM_000274.3(OAT):c.994G>A (p.Val332Met)	OAT	10	124401746	Gyrate atrophy of choroid and retina with pyridoxine-responsive ornithinemia;Ornithine aminotransferase deficiency
NM_024408.3(NOTCH2):c.6949C>T (p.Gln2317Ter)	NOTCH2	1	119915773	Hajdu-Cheney syndrome
NM_024408.3(NOTCH2):c.6622C>T (p.Gln2208Ter)	NOTCH2	1	119916100	Hajdu-Cheney syndrome
NM_024408.3(NOTCH2):c.7165C>T (p.Gln2389Ter)	NOTCH2	1	119915557	Hajdu-Cheney syndrome
NM_000522.4(HOXA13):c.1107G>A (p.Trp369Ter)	HOXA13	7	27198258	Hand foot uterus syndrome
NM_000334.4(SCN4A):c.2095G>A (p.Ala699Thr)	SCN4A	17	63957443	Handgrip myotonia;Myotonia

NM_001170535.2(ATAD3A):c.1582C>T (p.Arg528Trp)	ATAD3A	1	1529299	HAREL-YOON SYNDROME
NM_001170535.2(ATAD3A):c.158C>T (p.Thr53Ile)	ATAD3A	1	1512426	HAREL-YOON SYNDROME
NM_023110.2(FGFR1):c.2174G>A (p.Cys725Tyr)	FGFR1	8	38414164	Hartsfield syndrome
NM_000518.4(HBB):c.19G>A (p.Glu7Lys)	HBB	11	5227003	Hb SS disease;Malaria, resistance to;not provided
NM_000518.4(HBB):c.364G>A (p.Glu122Lys)	HBB	11	5225678	Hb SS disease;Sickle cell-Hemoglobin O Arab disease;beta Thalassemia
NM_004004.5(GJB2):c.158G>A (p.Cys53Tyr)	GJB2	13	20189424	Hearing impairment
NM_004004.5(GJB2):c.132G>A (p.Trp44Ter)	GJB2	13	20189450	Hearing impairment
NM_198159.2(MITF):c.943C>T (p.Arg315Ter)	MITF	3	69956460	Hearing impairment;Heterochromia iridis;Poliosis;Prelingual sensorineural hearing impairment;Waardenburg syndrome type 2A
NM_000287.3(PEX6):c.821C>T (p.Pro274Leu)	PEX6	6	42978330	Heimler syndrome 2
NM_000287.3(PEX6):c.1930C>T (p.Arg644Trp)	PEX6	6	42966813	Heimler syndrome 2
NM_000287.3(PEX6):c.1715C>T (p.Thr572Ile)	PEX6	6	42967537	Heimler syndrome 2
NM_000518.4(HBB):c.295G>A (p.Val99Met)	HBB	11	5226597	Heinz body anemia;Hemoglobinopathy
NM_002253.2(KDR):c.3439C>T (p.Pro1147Ser)	KDR	4	55038939	Hemangioma, capillary infantile;not specified
NM_001987.4(ETV6):c.641C>T (p.Pro214Leu)	ETV6	12	11869601	Hematologic neoplasm;Thrombocytopenia;Thrombocytopenia 5
NM_001987.4(ETV6):c.1106G>A (p.Arg369Gln)	ETV6	12	11884541	Hematologic neoplasm;Thrombocytopenia;Thrombocytopenia 5
NM_001987.4(ETV6):c.1195C>T (p.Arg399Cys)	ETV6	12	11885968	Hematologic neoplasm;Thrombocytopenia;Thrombocytopenia 5
NM_000410.3(HFE):c.506G>A (p.Trp169Ter)	HFE	6	26091479	Hemochromatosis type 1
NM_213653.3(HFE2):c.976C>T (p.Arg326Ter)	HFE2	1	146018382	Hemochromatosis type 2A
NM_021175.3(HAMP):c.166C>T (p.Arg56Ter)	HAMP	19	35284953	Hemochromatosis type 2B
NM_003227.3(TFR2):c.1186C>T (p.Arg396Ter)	TFR2	7	100630973	Hemochromatosis type 3
NM_003227.3(TFR2):c.1330G>A (p.Ala444Thr)	TFR2	7	100629313	Hemochromatosis type 3
NM_003227.3(TFR2):c.2137-1G>A	TFR2	7	100621127	Hemochromatosis type 3
NM_003227.3(TFR2):c.2374G>A (p.Gly792Arg)	TFR2	7	100620889	Hemochromatosis type 3
NM_003227.3(TFR2):c.313C>T (p.Arg105Ter)	TFR2	7	100640846	Hemochromatosis type 3
NM_003227.3(TFR2):c.64G>A (p.Val22Ile)	TFR2	7	100641198	Hemochromatosis type 3
NM_003227.3(TFR2):c.949C>T (p.Gln317Ter)	TFR2	7	100632099	Hemochromatosis type 3
NM_014585.5(SLC40A1):c.800G>A (p.Gly267Asp)	SLC40A1	2	189564186	Hemochromatosis type 4
NM_014585.5(SLC40A1):c.1469G>A (p.Gly490Asp)	SLC40A1	2	189562125	Hemochromatosis type 4
NM_000517.4(HBA2):c.314G>A (p.Cys105Tyr)	HBA2	16	173485	Hemoglobin H disease, nondeletional
NM_000517.4(HBA2):c.301-1G>A	HBA2	16	173471	Hemoglobin H disease, nondeletional
NM_000558.3(HBA1):c.179G>A (p.Gly60Asp)	HBA1	16	177012	Hemoglobin H disease, nondeletional
NM_000175.4(GPI):c.475G>A (p.Gly159Ser)	GPI	19	34377575	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
NM_000175.4(GPI):c.1040G>A (p.Arg347His)	GPI	19	34394044	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
NM_000175.4(GPI):c.1615G>A (p.Asp539Asn)	GPI	19	34399974	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
NM_000175.4(GPI):c.671C>T (p.Thr224Met)	GPI	19	34378971	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
NM_000175.4(GPI):c.14C>T (p.Thr5Ile)	GPI	19	34365280	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency
NM_001083116.1(PRF1):c.190C>T (p.Gln64Ter)	PRF1	10	70600713	Hemophagocytic lymphohistiocytosis, familial, 2
NM_001083116.1(PRF1):c.1286G>A (p.Gly429Glu)	PRF1	10	70598435	Hemophagocytic lymphohistiocytosis, familial, 2
NM_001083116.1(PRF1):c.1034C>T (p.Pro345Leu)	PRF1	10	70598687	Hemophagocytic lymphohistiocytosis, familial, 2
NM_001083116.1(PRF1):c.836G>A (p.Cys279Tyr)	PRF1	10	70598885	Hemophagocytic lymphohistiocytosis, familial, 2
NM_001083116.1(PRF1):c.1304C>T (p.Thr435Met)	PRF1	10	70598417	Hemophagocytic lymphohistiocytosis, familial, 2
NM_001083116.1(PRF1):c.1246C>T (p.Gln416Ter)	PRF1	10	70598475	Hemophagocytic lymphohistiocytosis, familial, 2
NM_001083116.1(PRF1):c.1122G>A (p.Trp374Ter)	PRF1	10	70598599	Hemophagocytic lymphohistiocytosis, familial, 2;Malignant lymphoma, non-Hodgkin
NM_199242.2(UNC13D):c.766C>T (p.Arg256Ter)	UNC13D	17	75840317	Hemophagocytic lymphohistiocytosis, familial, 3

NM_003764.3(STX11):c.802C>T (p.Gln268Ter)	STX11	6	144187429	Hemophagocytic lymphohistiocytosis, familial, 4
NM_006949.3(STXBP2):c.1430C>T (p.Pro477Leu)	STXBP2	19	7646322	Hemophagocytic lymphohistiocytosis, familial, 5
NM_006949.3(STXBP2):c.1621G>A (p.Gly541Ser)	STXBP2	19	7647436	Hemophagocytic lymphohistiocytosis, familial, 5
NM_000133.3(F9):c.677G>A (p.Arg226Gln)	F9	X	139551218	Hemophilia b(m)
NM_001845.5(COL4A1):c.1055C>T (p.Pro352Leu)	COL4A1	13	110201467	Hemorrhage, intracerebral, susceptibility to
NM_032801.4(JAM3):c.656G>A (p.Cys219Tyr)	JAM3	11	134145989	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts
NM_032801.4(JAM3):c.346G>A (p.Glu116Lys)	JAM3	11	134144330	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts
NM_133459.3(CCBE1):c.472C>T (p.Arg158Cys)	CCBE1	18	59466820	Hennekam lymphangiectasia-lymphedema syndrome
NM_001291303.1(FAT4):c.12851C>T (p.Ser4284Phe)	FAT4	4	125487373	Hennekam lymphangiectasia-lymphedema syndrome 2
NM_024582.4(FAT4):c.7123G>A (p.Glu2375Lys)	FAT4	4	125434349	Hennekam lymphangiectasia-lymphedema syndrome 2;Van Maldergem syndrome 2
NM_000185.3(SERPIND1):c.623G>A (p.Arg208His)	SERPIND1	22	20779935	Heparin cofactor ii deficiency
NM_000185.3(SERPIND1):c.1385C>T (p.Pro462Leu)	SERPIND1	22	20786951	Heparin cofactor ii deficiency
NM_000236.2(LIPC):c.1214C>T (p.Thr405Met)	LIPC	15	58563549	Hepatic lipase deficiency
NM_000429.2(MAT1A):c.790C>T (p.Arg264Cys)	MAT1A	10	80275178	Hepatic methionine adenosyltransferase deficiency
NM_000429.2(MAT1A):c.1006G>A (p.Gly336Arg)	MAT1A	10	80274599	Hepatic methionine adenosyltransferase deficiency
NM_000429.2(MAT1A):c.1070C>T (p.Pro357Leu)	MAT1A	10	80274535	Hepatic methionine adenosyltransferase deficiency;not provided
NM_000429.2(MAT1A):c.791G>A (p.Arg264His)	MAT1A	10	80275177	Hepatic methionine adenosyltransferase deficiency;not provided
NM_000876.3(IGF2R):c.4391G>A (p.Gly1464Glu)	IGF2R	6	160070006	Hepatocellular carcinoma
NM_000374.4(UROD):c.499G>A (p.Glu167Lys)	UROD	1	45013933	Hepatoerythropoietic porphyria
NM_000374.4(UROD):c.185C>T (p.Pro62Leu)	UROD	1	45013187	Hepatoerythropoietic porphyria
NM_130849.3(SLC39A4):c.599C>T (p.Pro200Leu)	SLC39A4	8	144415295	Hereditary acrodermatitis enteropathica
NM_130849.3(SLC39A4):c.1576G>A (p.Gly526Arg)	SLC39A4	8	144413288	Hereditary acrodermatitis enteropathica
NM_130849.3(SLC39A4):c.1120G>A (p.Gly374Arg)	SLC39A4	8	144414291	Hereditary acrodermatitis enteropathica
NM_130849.3(SLC39A4):c.989G>A (p.Gly330Asp)	SLC39A4	8	144414422	Hereditary acrodermatitis enteropathica
NM_130849.3(SLC39A4):c.283C>T (p.Arg95Cys)	SLC39A4	8	144416001	Hereditary acrodermatitis enteropathica
NM_000059.3(BRCA2):c.7007+1G>A	BRCA2	13	32346897	Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.7722G>A (p.Trp2574Ter)	BRCA2	13	32357846	Hereditary breast and ovarian cancer syndrome
NM_000059.3(BRCA2):c.2590C>T (p.Gln864Ter)	BRCA2	13	32336945	Hereditary breast and ovarian cancer syndrome
NM_000062.2(SERPING1):c.1372G>A (p.Ala458Thr)	SERPING1	11	57614450	Hereditary C1 esterase inhibitor deficiency - dysfunctional factor
NM_000062.2(SERPING1):c.1397G>A (p.Arg466His)	SERPING1	11	57614475	Hereditary C1 esterase inhibitor deficiency - dysfunctional factor;not provided
NM_000062.2(SERPING1):c.1396C>T (p.Arg466Cys)	SERPING1	11	57614474	Hereditary C1 esterase inhibitor deficiency - dysfunctional factor;not provided
NM_005732.3(RAD50):c.2821C>T (p.Gln941Ter)	RAD50	5	132608717	Hereditary cancer-predisposing syndrome
NM_001128425.1(MUTYH):c.393G>A (p.Trp131Ter)	MUTYH	1	45333166	Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.8473C>T (p.Gln2825Ter)	ATM	11	108345797	Hereditary cancer-predisposing syndrome
NM_005359.5(SMAD4):c.1345C>T (p.Gln449Ter)	SMAD4	18	51076674	Hereditary cancer-predisposing syndrome
NM_005591.3(MRE11):c.1090C>T (p.Arg364Ter)	MRE11	11	94467821	Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.283C>T (p.Gln95Ter)	ATM	11	108229275	Hereditary cancer-predisposing syndrome
NM_005732.3(RAD50):c.643C>T (p.Gln215Ter)	RAD50	5	132579953	Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.8998C>T (p.Gln3000Ter)	ATM	11	108365335	Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.6326G>A (p.Trp2109Ter)	ATM	11	108317500	Hereditary cancer-predisposing syndrome
NM_005591.3(MRE11):c.508C>T (p.Gln170Ter)	MRE11	11	94478771	Hereditary cancer-predisposing syndrome
NM_024675.3(PALB2):c.3476G>A (p.Trp1159Ter)	PALB2	16	23603544	Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.3077G>A (p.Trp1026Ter)	ATM	11	108271406	Hereditary cancer-predisposing syndrome
NM_003000.2(SDHB):c.575G>A (p.Cys192Tyr)	SDHB	1	17024040	Hereditary cancer-predisposing syndrome
NM_000546.5(TP53):c.994-1G>A	TP53	17	7670716	Hereditary cancer-predisposing syndrome

NM_024675.3(PALB2):c.1258C>T (p.Gln420Ter)	PALB2	16	23635288	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.4216C>T (p.Gln1406Ter)	APC	5	112839810	Hereditary cancer-predisposing syndrome
NM_032043.2(BRIP1):c.2576-1G>A	BRIP1	17	61686166	Hereditary cancer-predisposing syndrome
NM_032043.2(BRIP1):c.2377C>T (p.Gln793Ter)	BRIP1	17	61743015	Hereditary cancer-predisposing syndrome
NM_000314.6(PTEN):c.822G>A (p.Trp274Ter)	PTEN	10	87960914	Hereditary cancer-predisposing syndrome
NM_004360.4(CDH1):c.1921C>T (p.Gln641Ter)	CDH1	16	68822210	Hereditary cancer-predisposing syndrome
NM_004360.4(CDH1):c.1147C>T (p.Gln383Ter)	CDH1	16	68813322	Hereditary cancer-predisposing syndrome
NM_003000.2(SDHB):c.688C>T (p.Arg230Cys)	SDHB	1	17022685	Hereditary cancer-predisposing syndrome
NM_000465.3(BARD1):c.2268G>A (p.Trp756Ter)	BARD1	2	214728742	Hereditary cancer-predisposing syndrome
NM_000179.2(MSH6):c.1238G>A (p.Trp413Ter)	MSH6	2	47799221	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.4621C>T (p.Gln1541Ter)	APC	5	112840215	Hereditary cancer-predisposing syndrome
NM_005732.3(RAD50):c.412C>T (p.Arg138Ter)	RAD50	5	132579363	Hereditary cancer-predisposing syndrome
NM_005732.3(RAD50):c.1237C>T (p.Gln413Ter)	RAD50	5	132588872	Hereditary cancer-predisposing syndrome
NM_005732.3(RAD50):c.3598C>T (p.Arg1200Ter)	RAD50	5	132638203	Hereditary cancer-predisposing syndrome
NM_000535.6(PMS2):c.1411C>T (p.Gln471Ter)	PMS2	7	5987354	Hereditary cancer-predisposing syndrome
NM_000535.6(PMS2):c.354-1G>A	PMS2	7	6002637	Hereditary cancer-predisposing syndrome
NM_000314.6(PTEN):c.733C>T (p.Gln245Ter)	PTEN	10	87957951	Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.151C>T (p.Gln51Ter)	ATM	11	108227854	Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.1737G>A (p.Trp579Ter)	ATM	11	108251966	Hereditary cancer-predisposing syndrome
NM_000051.3(ATM):c.3349C>T (p.Gln1117Ter)	ATM	11	108279555	Hereditary cancer-predisposing syndrome
NM_005591.3(MRE11):c.1726C>T (p.Arg576Ter)	MRE11	11	94447276	Hereditary cancer-predisposing syndrome
NM_002382.4(MAX):c.295+1G>A	MAX	14	65077912	Hereditary cancer-predisposing syndrome
NM_004360.4(CDH1):c.60G>A (p.Trp20Ter)	CDH1	16	68738308	Hereditary cancer-predisposing syndrome
NM_001042492.2(NF1):c.3208C>T (p.Gln1070Ter)	NF1	17	31232083	Hereditary cancer-predisposing syndrome
NM_001042492.2(NF1):c.8095C>T (p.Gln2699Ter)	NF1	17	31358604	Hereditary cancer-predisposing syndrome
NM_007294.3(BRCA1):c.4185+1G>A	BRCA1	17	43090943	Hereditary cancer-predisposing syndrome
NM_032043.2(BRIP1):c.1005G>A (p.Trp335Ter)	BRIP1	17	61801388	Hereditary cancer-predisposing syndrome
NM_032043.2(BRIP1):c.667C>T (p.Gln223Ter)	BRIP1	17	61808718	Hereditary cancer-predisposing syndrome
NM_007194.3(CHEK2):c.278G>A (p.Trp93Ter)	CHEK2	22	28734444	Hereditary cancer-predisposing syndrome
NM_004656.3(BAP1):c.1063C>T (p.Gln355Ter)	BAP1	3	52405163	Hereditary cancer-predisposing syndrome
NM_003000.2(SDHB):c.445C>T (p.Gln149Ter)	SDHB	1	17027844	Hereditary cancer-predisposing syndrome
NM_001128425.1(MUTYH):c.421C>T (p.Gln141Ter)	MUTYH	1	45333138	Hereditary cancer-predisposing syndrome
NM_000465.3(BARD1):c.1216C>T (p.Arg406Ter)	BARD1	2	214780658	Hereditary cancer-predisposing syndrome
NM_000465.3(BARD1):c.277C>T (p.Gln93Ter)	BARD1	2	214792384	Hereditary cancer-predisposing syndrome
NM_000179.2(MSH6):c.2331G>A (p.Trp777Ter)	MSH6	2	47800314	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.933+1G>A	APC	5	112815594	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.1234C>T (p.Gln412Ter)	APC	5	112819266	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.1417C>T (p.Gln473Ter)	APC	5	112827116	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.1873C>T (p.Gln625Ter)	APC	5	112835080	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.1999C>T (p.Gln667Ter)	APC	5	112837593	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.3146G>A (p.Trp1049Ter)	APC	5	112838740	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.3211C>T (p.Gln1071Ter)	APC	5	112838805	Hereditary cancer-predisposing syndrome
NM_000038.5(APC):c.5038C>T (p.Gln1680Ter)	APC	5	112840632	Hereditary cancer-predisposing syndrome
NM_005732.3(RAD50):c.2284C>T (p.Gln762Ter)	RAD50	5	132603376	Hereditary cancer-predisposing syndrome
NM_005732.3(RAD50):c.3229C>T (p.Arg1077Ter)	RAD50	5	132618134	Hereditary cancer-predisposing syndrome

NM_005732.3(RAD50):c.3454C>T (p.Arg1152Ter)	RAD50	5	132637179	Hereditary cancer-predisposing syndrome	
NM_005732.3(RAD50):c.3787C>T (p.Gln1263Ter)	RAD50	5	132642212	Hereditary cancer-predisposing syndrome	
NM_000535.6(PMS2):c.2137C>T (p.Gln713Ter)	PMS2	7	5982861	Hereditary cancer-predisposing syndrome	
NM_000535.6(PMS2):c.1198C>T (p.Gln400Ter)	PMS2	7	5987567	Hereditary cancer-predisposing syndrome	
NM_000077.4(CDKN2A):c.44G>A (p.Trp15Ter)	CDKN2A	9	21974784	Hereditary cancer-predisposing syndrome	
NM_000051.3(ATM):c.67C>T (p.Arg23Ter)	ATM	11	108227691	Hereditary cancer-predisposing syndrome	
NM_000051.3(ATM):c.205C>T (p.Gln69Ter)	ATM	11	108229197	Hereditary cancer-predisposing syndrome	
NM_000051.3(ATM):c.3511C>T (p.Gln1171Ter)	ATM	11	108281103	Hereditary cancer-predisposing syndrome	
NM_000051.3(ATM):c.6082C>T (p.Gln2028Ter)	ATM	11	108315898	Hereditary cancer-predisposing syndrome	
NM_000051.3(ATM):c.7189C>T (p.Gln2397Ter)	ATM	11	108329120	Hereditary cancer-predisposing syndrome	
NM_000051.3(ATM):c.8218C>T (p.Gln2740Ter)	ATM	11	108335911	Hereditary cancer-predisposing syndrome	
NM_000051.3(ATM):c.8287C>T (p.Arg2763Ter)	ATM	11	108343240	Hereditary cancer-predisposing syndrome	
NM_005591.3(MRE11):c.1447C>T (p.Arg483Ter)	MRE11	11	94459461	Hereditary cancer-predisposing syndrome	
NM_024675.3(PALB2):c.3114G>A (p.Trp1038Ter)	PALB2	16	23614091	Hereditary cancer-predisposing syndrome	
NM_024675.3(PALB2):c.2630G>A (p.Trp877Ter)	PALB2	16	23626354	Hereditary cancer-predisposing syndrome	
NM_024675.3(PALB2):c.1378C>T (p.Gln460Ter)	PALB2	16	23635168	Hereditary cancer-predisposing syndrome	
NM_024675.3(PALB2):c.115C>T (p.Gln39Ter)	PALB2	16	23637946	Hereditary cancer-predisposing syndrome	
NM_004360.4(CDH1):c.2293C>T (p.Gln765Ter)	CDH1	16	68828302	Hereditary cancer-predisposing syndrome	
NM_001042492.2(NF1):c.2887C>T (p.Gln963Ter)	NF1	17	31229871	Hereditary cancer-predisposing syndrome	
NM_001042492.2(NF1):c.3941G>A (p.Trp1314Ter)	NF1	17	31235988	Hereditary cancer-predisposing syndrome	
NM_001042492.2(NF1):c.5269-1G>A	NF1	17	31327498	Hereditary cancer-predisposing syndrome	
NM_001042492.2(NF1):c.5812+1G>A	NF1	17	31330499	Hereditary cancer-predisposing syndrome	
NM_001042492.2(NF1):c.5991G>A (p.Trp1997Ter)	NF1	17	31335016	Hereditary cancer-predisposing syndrome	
NM_002878.3(RAD51D):c.442C>T (p.Gln148Ter)	RAD51D	17	35107026	Hereditary cancer-predisposing syndrome	
NM_007294.3(BRCA1):c.4162C>T (p.Gln1388Ter)	BRCA1	17	43090967	Hereditary cancer-predisposing syndrome	
NM_007294.3(BRCA1):c.3979C>T (p.Gln1327Ter)	BRCA1	17	43091552	Hereditary cancer-predisposing syndrome	
NM_032043.2(BRIP1):c.2053C>T (p.Gln685Ter)	BRIP1	17	61776445	Hereditary cancer-predisposing syndrome	
NM_007194.3(CHEK2):c.1459C>T (p.Gln487Ter)	CHEK2	22	28694034	Hereditary cancer-predisposing syndrome	
NM_000546.5(TP53):c.158G>A (p.Trp53Ter)	TP53	17	7676211	Hereditary cancer-predisposing syndrome	
NM_005359.5(SMAD4):c.297G>A (p.Trp99Ter)	SMAD4	18	51048733	Hereditary cancer-predisposing syndrome	
NM_005359.5(SMAD4):c.931C>T (p.Gln311Ter)	SMAD4	18	51059892	Hereditary cancer-predisposing syndrome	
NM_005732.3(RAD50):c.1966C>T (p.Arg656Ter)	RAD50	5	132595041	Hereditary cancer-predisposing syndrome	
NM_024675.3(PALB2):c.106C>T (p.Gln36Ter)	PALB2	16	23638072	Hereditary cancer-predisposing syndrome	
NM_005732.3(RAD50):c.832C>T (p.Arg278Ter)	RAD50	5	132587637	Hereditary cancer-predisposing syndrome	
NM_005732.3(RAD50):c.2467C>T (p.Arg823Ter)	RAD50	5	132603989	Hereditary cancer-predisposing syndrome	
NM_005732.3(RAD50):c.2569C>T (p.Gln857Ter)	RAD50	5	132604850	Hereditary cancer-predisposing syndrome	
NM_000077.4(CDKN2A):c.148C>T (p.Gln50Ter)	CDKN2A	9	21974680	Hereditary cancer-predisposing syndrome;Hereditary cutaneous melanoma;not provided	
NM_004360.4(CDH1):c.1137G>A (p.Thr379=)	CDH1	16	68812263	Hereditary cancer-predisposing syndrome;Hereditary diffuse gastric cancer	
NM_004360.4(CDH1):c.1792C>T (p.Arg598Ter)	CDH1	16	68822081	Hereditary cancer-predisposing syndrome;Hereditary diffuse gastric cancer;not provided	
NM_004360.4(CDH1):c.1565+1G>A	CDH1	16	68815760	Hereditary cancer-predisposing syndrome;Hereditary diffuse gastric cancer;not provided	
NM_004360.4(CDH1):c.1003C>T (p.Arg335Ter)	CDH1	16	68811854	Hereditary cancer-predisposing syndrome;Hereditary diffuse gastric cancer;not provided	
NM_004360.4(CDH1):c.1137+1G>A	CDH1	16	68812264	Hereditary cancer-predisposing syndrome;Hereditary diffuse gastric cancer;not provided	
NM_000143.3(FH):c.301C>T (p.Arg101Ter)	FH	1	241513680	Hereditary cancer-predisposing syndrome;Hereditary leiomyomatosis and renal cell cancer;not provided	

NM_000143.3(FH):c.1027C>T (p.Arg343Ter)	FH	1	241504123	Hereditary cancer-predisposing syndrome;Hereditary leiomyomatosis and renal cell cancer;not provided
NM_000143.3(FH):c.697C>T (p.Arg233Cys)	FH	1	241508644	Hereditary cancer-predisposing syndrome;Hereditary leiomyomatosis and renal cell cancer;not provided
NM_000535.6(PMS2):c.1882C>T (p.Arg628Ter)	PMS2	7	5986883	Hereditary cancer-predisposing syndrome;Hereditary nonpolyposis colorectal cancer type 4;Lynch syndrome;not provided
NM_000535.6(PMS2):c.2174+1G>A	PMS2	7	5982823	Hereditary cancer-predisposing syndrome;Hereditary nonpolyposis colorectal cancer type 4;Lynch syndrome;not provided
NM_000535.6(PMS2):c.2404C>T (p.Arg802Ter)	PMS2	7	5977629	Hereditary cancer-predisposing syndrome;Hereditary nonpolyposis colorectal cancer type 4;Lynch syndrome;Turcot syndrome;not provided
NM_000179.2(MSH6):c.3991C>T (p.Arg1331Ter)	MSH6	2	47806641	Hereditary cancer-predisposing syndrome;Hereditary nonpolyposis colorectal cancer type 5;Lynch syndrome;not provided
NM_000179.2(MSH6):c.10C>T (p.Gln4Ter)	MSH6	2	47783243	Hereditary cancer-predisposing syndrome;Hereditary nonpolyposis colorectal cancer type 5;Lynch syndrome;not provided
NM_004329.2(BMPR1A):c.682C>T (p.Arg228Ter)	BMPR1A	10	86917140	Hereditary cancer-predisposing syndrome;Juvenile polyposis syndrome
NM_004329.2(BMPR1A):c.1081C>T (p.Arg361Ter)	BMPR1A	10	86919384	Hereditary cancer-predisposing syndrome;Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.1333C>T (p.Arg445Ter)	SMAD4	18	51076662	Hereditary cancer-predisposing syndrome;Juvenile polyposis syndrome;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
NM_004329.2(BMPR1A):c.817C>T (p.Arg273Ter)	BMPR1A	10	86917275	Hereditary cancer-predisposing syndrome;Juvenile polyposis syndrome;not provided
NM_000546.5(TP53):c.1009C>T (p.Arg337Cys)	TP53	17	7670700	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome
NM_000546.5(TP53):c.455C>T (p.Pro152Leu)	TP53	17	7675157	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome
NM_000546.5(TP53):c.273G>A (p.Trp91Ter)	TP53	17	7676096	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome
NM_000546.5(TP53):c.524G>A (p.Arg175His)	TP53	17	7675088	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni syndrome 1;Malignant tumor of esophagus;Neoplasm;Neoplasm of breast;not provided
NM_000546.5(TP53):c.637C>T (p.Arg213Ter)	TP53	17	7674894	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;Li-Fraumeni syndrome 1;not provided
NM_000546.5(TP53):c.586C>T (p.Arg196Ter)	TP53	17	7674945	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;not provided
NM_000546.5(TP53):c.473G>A (p.Arg158His)	TP53	17	7675139	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;not provided
NM_000546.5(TP53):c.542G>A (p.Arg181His)	TP53	17	7675070	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;not provided
NM_000546.5(TP53):c.375G>A (p.Thr125=)	TP53	17	7675994	Hereditary cancer-predisposing syndrome;Li-Fraumeni syndrome;not provided
NM_001042492.2(NF1):c.7348C>T (p.Arg2450Ter)	NF1	17	31350209	Hereditary cancer-predisposing syndrome;Lisch nodules;Subcutaneous neurofibromas;not provided
NM_000179.2(MSH6):c.2503C>T (p.Gln835Ter)	MSH6	2	47800486	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000179.2(MSH6):c.718C>T (p.Arg240Ter)	MSH6	2	47798701	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000249.3(MLH1):c.1038G>A (p.Gln346=)	MLH1	3	37020463	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000249.3(MLH1):c.245C>T (p.Thr82Ile)	MLH1	3	37000992	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000249.3(MLH1):c.83C>T (p.Pro28Leu)	MLH1	3	36993630	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000249.3(MLH1):c.884G>A (p.Ser295Asn)	MLH1	3	37017599	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000249.3(MLH1):c.982C>T (p.Gln328Ter)	MLH1	3	37020407	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000251.2(MSH2):c.1046C>T (p.Pro349Leu)	MSH2	2	47416399	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000251.2(MSH2):c.1120C>T (p.Gln374Ter)	MSH2	2	47429785	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000251.2(MSH2):c.1285C>T (p.Gln429Ter)	MSH2	2	47445556	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000251.2(MSH2):c.1477C>T (p.Gln493Ter)	MSH2	2	47463121	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000251.2(MSH2):c.2152C>T (p.Gln718Ter)	MSH2	2	47476513	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000535.6(PMS2):c.703C>T (p.Gln235Ter)	PMS2	7	5999110	Hereditary cancer-predisposing syndrome;Lynch syndrome
NM_000535.6(PMS2):c.88C>T (p.Gln30Ter)	PMS2	7	6005967	Hereditary cancer-predisposing syndrome;Lynch syndrome

NM_000249.3(MLH1):c.2141G>A (p.Trp714Ter)	MLH1	3	37050523	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome I
NM_000251.2(MSH2):c.1009C>T (p.Gln337Ter)	MSH2	2	47416362	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome I
NM_000251.2(MSH2):c.1216C>T (p.Arg406Ter)	MSH2	2	47429881	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome I;not provided
NM_000179.2(MSH6):c.3202C>T (p.Arg1068Ter)	MSH6	2	47803449	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome I;not provided
NM_000251.2(MSH2):c.1165C>T (p.Arg389Ter)	MSH2	2	47429830	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome I;not provided
NM_000249.3(MLH1):c.1943C>T (p.Pro648Leu)	MLH1	3	37048563	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome II;Muir-TorrÃ© syndrome;Turcot syndrome
NM_000249.3(MLH1):c.200G>A (p.Gly67Glu)	MLH1	3	36996702	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome II;not provided
NM_000249.3(MLH1):c.793C>T (p.Arg265Cys)	MLH1	3	37017508	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome II;not provided
NM_000249.3(MLH1):c.2059C>T (p.Arg687Trp)	MLH1	3	37048973	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome II;not provided
NM_000249.3(MLH1):c.677G>A (p.Arg226Gln)	MLH1	3	37012099	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome II;not provided
NM_000249.3(MLH1):c.2041G>A (p.Ala681Thr)	MLH1	3	37048955	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome II;not specified
NM_000249.3(MLH1):c.1942C>T (p.Pro648Ser)	MLH1	3	37048562	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome II;Turcot syndrome
NM_000249.3(MLH1):c.676C>T (p.Arg226Ter)	MLH1	3	37012098	Hereditary cancer-predisposing syndrome;Lynch syndrome;Lynch syndrome II;Turcot syndrome;not provided
NM_000251.2(MSH2):c.2038C>T (p.Arg680Ter)	MSH2	2	47476399	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000179.2(MSH6):c.1483C>T (p.Arg495Ter)	MSH6	2	47799466	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000179.2(MSH6):c.2194C>T (p.Arg732Ter)	MSH6	2	47800177	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000179.2(MSH6):c.3103C>T (p.Arg1035Ter)	MSH6	2	47801086	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000179.2(MSH6):c.892C>T (p.Arg298Ter)	MSH6	2	47798875	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000249.3(MLH1):c.1459C>T (p.Arg487Ter)	MLH1	3	37028833	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000249.3(MLH1):c.1731G>A (p.Ser577=)	MLH1	3	37042331	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000249.3(MLH1):c.184C>T (p.Gln62Ter)	MLH1	3	36996686	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000249.3(MLH1):c.1975C>T (p.Arg659Ter)	MLH1	3	37048595	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000249.3(MLH1):c.199G>A (p.Gly67Arg)	MLH1	3	36996701	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000251.2(MSH2):c.1076+1G>A	MSH2	2	47416430	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000251.2(MSH2):c.1147C>T (p.Arg383Ter)	MSH2	2	47429812	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000251.2(MSH2):c.181C>T (p.Gln61Ter)	MSH2	2	47403372	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000251.2(MSH2):c.212-1G>A	MSH2	2	47408400	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000251.2(MSH2):c.2131C>T (p.Arg711Ter)	MSH2	2	47476492	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000535.6(PMS2):c.697C>T (p.Gln233Ter)	PMS2	7	5999116	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided
NM_000535.6(PMS2):c.1261C>T (p.Arg421Ter)	PMS2	7	5987504	Hereditary cancer-predisposing syndrome;Lynch syndrome;not provided;not specified
NM_000251.2(MSH2):c.2047G>A (p.Gly683Arg)	MSH2	2	47476408	Hereditary cancer-predisposing syndrome;Lynch syndrome;not specified
NM_000535.6(PMS2):c.400C>T (p.Arg134Ter)	PMS2	7	6002590	Hereditary cancer-predisposing syndrome;Lynch syndrome;Turcot syndrome;not provided
NM_000535.6(PMS2):c.1927C>T (p.Gln643Ter)	PMS2	7	5986838	Hereditary cancer-predisposing syndrome;Lynch syndrome;Turcot syndrome;not provided
NM_020975.4(RET):c.1853G>A (p.Cys618Tyr)	RET	10	43113649	Hereditary cancer-predisposing syndrome;Medullary thyroid carcinoma;Multiple endocrine neoplasia, type 1;Multiple endocrine neoplasia, type 2;Multiple endocrine neoplasia, type 2a;Multiple endocrine neoplasia, type 2b;Multiple endocrine neoplasia, type 4
NM_020975.4(RET):c.1901G>A (p.Cys634Tyr)	RET	10	43114501	Hereditary cancer-predisposing syndrome;Medullary thyroid carcinoma;Multiple endocrine neoplasia, type 1;Multiple endocrine neoplasia, type 2;Multiple endocrine neoplasia, type 2a;Multiple endocrine neoplasia, type 2b;Multiple endocrine neoplasia, type 4;Neoplasm;Pheochromocytoma;not provided
NM_002485.4(NBN):c.976C>T (p.Gln326Ter)	NBN	8	89964428	Hereditary cancer-predisposing syndrome;Microcephaly, normal intelligence and immunodeficiency
NM_002485.4(NBN):c.127C>T (p.Arg43Ter)	NBN	8	89982765	Hereditary cancer-predisposing syndrome;Microcephaly, normal intelligence and immunodeficiency
NM_002485.4(NBN):c.1747C>T (p.Gln583Ter)	NBN	8	89953342	Hereditary cancer-predisposing syndrome;Microcephaly, normal intelligence and immunodeficiency

NM_002485.4(NBN):c.2140C>T (p.Arg714Ter)	NBN	8	89943297	Hereditary cancer-predisposing syndrome;Microcephaly, normal intelligence and immunodeficiency;not provided
NM_002485.4(NBN):c.1030C>T (p.Gln344Ter)	NBN	8	89958819	Hereditary cancer-predisposing syndrome;Microcephaly, normal intelligence and immunodeficiency;not provided
NM_130799.2(MEN1):c.1378C>T (p.Arg460Ter)	MEN1	11	64804789	Hereditary cancer-predisposing syndrome;Multiple endocrine neoplasia, type 1;not provided
NM_144997.5(FLCN):c.250-1G>A	FLCN	17	17226323	Hereditary cancer-predisposing syndrome;Multiple fibrofolliculomas
NM_144997.5(FLCN):c.557G>A (p.Trp186Ter)	FLCN	17	17223983	Hereditary cancer-predisposing syndrome;Multiple fibrofolliculomas
NM_144997.5(FLCN):c.499C>T (p.Gln167Ter)	FLCN	17	17224041	Hereditary cancer-predisposing syndrome;Multiple fibrofolliculomas;not provided
NM_001128425.1(MUTYH):c.799C>T (p.Gln267Ter)	MUTYH	1	45332300	Hereditary cancer-predisposing syndrome;MYH-associated polyposis
NM_001128425.1(MUTYH):c.389-1G>A	MUTYH	1	45333171	Hereditary cancer-predisposing syndrome;MYH-associated polyposis
NM_001128425.1(MUTYH):c.309G>A (p.Trp103Ter)	MUTYH	1	45333452	Hereditary cancer-predisposing syndrome;MYH-associated polyposis
NM_001128425.1(MUTYH):c.733C>T (p.Arg245Cys)	MUTYH	1	45332446	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.1012C>T (p.Gln338Ter)	MUTYH	1	45331835	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.289C>T (p.Arg97Ter)	MUTYH	1	45333472	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.734G>A (p.Arg245His)	MUTYH	1	45332445	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.857G>A (p.Gly286Glu)	MUTYH	1	45332242	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.545G>A (p.Arg182His)	MUTYH	1	45332794	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.884C>T (p.Pro295Leu)	MUTYH	1	45332215	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.721C>T (p.Arg241Trp)	MUTYH	1	45332458	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.544C>T (p.Arg182Cys)	MUTYH	1	45332795	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.325C>T (p.Arg109Trp)	MUTYH	1	45333436	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_001128425.1(MUTYH):c.467G>A (p.Trp156Ter)	MUTYH	1	45332955	Hereditary cancer-predisposing syndrome;MYH-associated polyposis;not provided
NM_000314.6(PTEN):c.517C>T (p.Arg173Cys)	PTEN	10	87952142	Hereditary cancer-predisposing syndrome;Neoplasm of brain;not provided
NM_001042492.2(NF1):c.3721C>T (p.Arg1241Ter)	NF1	17	31235623	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1
NM_001042492.2(NF1):c.625C>T (p.Gln209Ter)	NF1	17	31181460	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1
NM_001042492.2(NF1):c.910C>T (p.Arg304Ter)	NF1	17	31200443	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1
NM_001042492.2(NF1):c.5609G>A (p.Arg1870Gln)	NF1	17	31327839	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1
NM_001042492.2(NF1):c.7909C>T (p.Arg2637Ter)	NF1	17	31357308	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1
NM_001042492.2(NF1):c.1318C>T (p.Arg440Ter)	NF1	17	31206297	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1
NM_001042492.2(NF1):c.6772C>T (p.Arg2258Ter)	NF1	17	31338092	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1
NM_001042492.2(NF1):c.7549C>T (p.Arg2517Ter)	NF1	17	31352348	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1
NM_001042492.2(NF1):c.1885G>A (p.Gly629Arg)	NF1	17	31225134	Hereditary cancer-predisposing syndrome;Neurofibromatosis, type 1;not provided
NM_005732.3(RAD50):c.3553C>T (p.Arg1185Ter)	RAD50	5	132638158	Hereditary cancer-predisposing syndrome;Nijmegen breakage syndrome-like disorder
NM_144997.5(FLCN):c.1117C>T (p.Gln373Ter)	FLCN	17	17217128	Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.3836G>A (p.Trp1279Ter)	ATM	11	108284316	Hereditary cancer-predisposing syndrome;not provided
NM_000535.6(PMS2):c.823C>T (p.Gln275Ter)	PMS2	7	5995614	Hereditary cancer-predisposing syndrome;not provided
NM_005732.3(RAD50):c.1393C>T (p.Gln465Ter)	RAD50	5	132589778	Hereditary cancer-predisposing syndrome;not provided
NM_032043.2(BRIP1):c.1315C>T (p.Arg439Ter)	BRIP1	17	61799125	Hereditary cancer-predisposing syndrome;not provided
NM_001128425.1(MUTYH):c.940C>T (p.Gln314Ter)	MUTYH	1	45332080	Hereditary cancer-predisposing syndrome;not provided
NM_000179.2(MSH6):c.2932C>T (p.Gln978Ter)	MSH6	2	47800915	Hereditary cancer-predisposing syndrome;not provided
NM_000465.3(BARD1):c.733C>T (p.Gln245Ter)	BARD1	2	214781141	Hereditary cancer-predisposing syndrome;not provided
NM_024675.3(PALB2):c.2712G>A (p.Trp904Ter)	PALB2	16	23626272	Hereditary cancer-predisposing syndrome;not provided
NM_000546.5(TP53):c.920-1G>A	TP53	17	7673609	Hereditary cancer-predisposing syndrome;not provided
NM_032043.2(BRIP1):c.463C>T (p.Gln155Ter)	BRIP1	17	61849173	Hereditary cancer-predisposing syndrome;not provided
NM_002485.4(NBN):c.1474C>T (p.Gln492Ter)	NBN	8	89953615	Hereditary cancer-predisposing syndrome;not provided

NM_000314.6(PTEN):c.737C>T (p.Pro246Leu)	PTEN	10	87957955	Hereditary cancer-predisposing syndrome;not provided
NM_032043.2(BRIP1):c.1066C>T (p.Arg356Ter)	BRIP1	17	61801327	Hereditary cancer-predisposing syndrome;not provided
NM_000546.5(TP53):c.1024C>T (p.Arg342Ter)	TP53	17	7670685	Hereditary cancer-predisposing syndrome;not provided
NM_000546.5(TP53):c.493C>T (p.Gln165Ter)	TP53	17	7675119	Hereditary cancer-predisposing syndrome;not provided
NM_000465.3(BARD1):c.496C>T (p.Gln166Ter)	BARD1	2	214781378	Hereditary cancer-predisposing syndrome;not provided
NM_000251.2(MSH2):c.1351C>T (p.Gln451Ter)	MSH2	2	47445622	Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.6709C>T (p.Arg2237Ter)	APC	5	112842303	Hereditary cancer-predisposing syndrome;not provided
NM_004329.2(BMPR1A):c.1480C>T (p.Arg494Ter)	BMPR1A	10	86923600	Hereditary cancer-predisposing syndrome;not provided
NM_000314.6(PTEN):c.165-1G>A	PTEN	10	87925512	Hereditary cancer-predisposing syndrome;not provided
NM_000314.6(PTEN):c.892C>T (p.Gln298Ter)	PTEN	10	87960984	Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.2098C>T (p.Gln700Ter)	ATM	11	108254013	Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.4852C>T (p.Arg1618Ter)	ATM	11	108295002	Hereditary cancer-predisposing syndrome;not provided
NM_000314.6(PTEN):c.407G>A (p.Cys136Tyr)	PTEN	10	87933166	Hereditary cancer-predisposing syndrome;not provided
NM_000038.5(APC):c.3147G>A (p.Trp1049Ter)	APC	5	112838741	Hereditary cancer-predisposing syndrome;not provided
NM_001042492.2(NF1):c.31C>T (p.Gln11Ter)	NF1	17	31095340	Hereditary cancer-predisposing syndrome;not provided
NM_007194.3(CHEK2):c.205C>T (p.Gln69Ter)	CHEK2	22	28734517	Hereditary cancer-predisposing syndrome;not provided
NM_001128425.1(MUTYH):c.628C>T (p.Gln210Ter)	MUTYH	1	45332636	Hereditary cancer-predisposing syndrome;not provided
NM_000465.3(BARD1):c.709C>T (p.Gln237Ter)	BARD1	2	214781165	Hereditary cancer-predisposing syndrome;not provided
NM_000535.6(PMS2):c.2155C>T (p.Gln719Ter)	PMS2	7	5982843	Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.332-1G>A	ATM	11	108235669	Hereditary cancer-predisposing syndrome;not provided
NM_000051.3(ATM):c.1396C>T (p.Gln466Ter)	ATM	11	108250861	Hereditary cancer-predisposing syndrome;not provided
NM_024675.3(PALB2):c.2693G>A (p.Trp898Ter)	PALB2	16	23626291	Hereditary cancer-predisposing syndrome;not provided
NM_058216.2(RAD51C):c.914G>A (p.Trp305Ter)	RAD51C	17	58724049	Hereditary cancer-predisposing syndrome;not provided
NM_007194.3(CHEK2):c.1315C>T (p.Gln439Ter)	CHEK2	22	28695187	Hereditary cancer-predisposing syndrome;not provided
NM_024675.3(PALB2):c.2368C>T (p.Gln790Ter)	PALB2	16	23629786	Hereditary cancer-predisposing syndrome;not provided
NM_000535.6(PMS2):c.1687C>T (p.Arg563Ter)	PMS2	7	5987078	Hereditary cancer-predisposing syndrome;not provided;not specified
NM_003002.3(SDHD):c.129G>A (p.Trp43Ter)	SDHD	11	112087933	Hereditary cancer-predisposing syndrome;Paragangliomas 1
NM_017841.2(SDHAF2):c.232G>A (p.Gly78Arg)	SDHAF2	11	61437820	Hereditary cancer-predisposing syndrome;Paragangliomas 2
NM_003001.3(SDHC):c.43C>T (p.Arg15Ter)	SDHC	1	161323636	Hereditary cancer-predisposing syndrome;Paragangliomas 3;not provided
NM_003001.3(SDHC):c.397C>T (p.Arg133Ter)	SDHC	1	161356832	Hereditary cancer-predisposing syndrome;Paragangliomas 3;not provided
NM_000455.4(STK11):c.910C>T (p.Arg304Trp)	STK11	19	1221996	Hereditary cancer-predisposing syndrome;Peutz-Jeghers syndrome;not provided
NM_002382.4(MAX):c.223C>T (p.Arg75Ter)	MAX	14	65077985	Hereditary cancer-predisposing syndrome;Pheochromocytoma, susceptibility to
NM_017849.3(TMEM127):c.475C>T (p.Gln159Ter)	TMEM127	2	96254050	Hereditary cancer-predisposing syndrome;Pheochromocytoma;Pheochromocytoma, susceptibility to
NM_000314.6(PTEN):c.49C>T (p.Gln17Ter)	PTEN	10	87864518	Hereditary cancer-predisposing syndrome;PTEN hamartoma tumor syndrome
NM_001127500.2(MET):c.3712G>A (p.Val1238Ile)	MET	7	116783329	Hereditary cancer-predisposing syndrome;Renal cell carcinoma, papillary, 1
NM_000548.4(TSC2):c.2714G>A (p.Arg905Gln)	TSC2	16	2076142	Hereditary cancer-predisposing syndrome;Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.5138G>A (p.Arg1713His)	TSC2	16	2088117	Hereditary cancer-predisposing syndrome;Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_002144.3(HOXB1):c.619C>T (p.Arg207Cys)	HOXB1	17	48529834	Hereditary congenital facial paresis 3
NM_000077.4(CDKN2A):c.-19311G>A	CDKN2A	9	21994138	Hereditary cutaneous melanoma;not provided
NM_004360.4(CDH1):c.2095C>T (p.Gln699Ter)	CDH1	16	68823557	Hereditary diffuse gastric cancer
NM_004360.4(CDH1):c.59G>A (p.Trp20Ter)	CDH1	16	68738307	Hereditary diffuse gastric cancer
NM_004360.4(CDH1):c.1901C>T (p.Ala634Val)	CDH1	16	68822190	Hereditary diffuse gastric cancer
NM_004360.4(CDH1):c.187C>T (p.Arg63Ter)	CDH1	16	68801693	Hereditary diffuse gastric cancer

NM_004360.4(CDH1):c.220C>T (p.Arg74Ter)	CDH1	16	68801726	Hereditary diffuse gastric cancer
NM_005211.3(CSF1R):c.1897G>A (p.Glu633Lys)	CSF1R	5	150060934	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.1766G>A (p.Gly589Glu)	CSF1R	5	150061583	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.2345G>A (p.Arg782His)	CSF1R	5	150056316	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.1958G>A (p.Cys653Tyr)	CSF1R	5	150060873	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.2329C>T (p.Arg777Trp)	CSF1R	5	150056332	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.2701C>T (p.Pro901Ser)	CSF1R	5	150054384	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.2629C>T (p.Gln877Ter)	CSF1R	5	150055262	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3:c.2467C>T	CSF1R	5	150056112	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.2350G>A (p.Val784Met)	CSF1R	5	150056311	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.2342C>T (p.Ala781Val)	CSF1R	5	150056319	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.2330G>A (p.Arg777Gln)	CSF1R	5	150056331	Hereditary diffuse leukoencephalopathy with spheroids
NM_005211.3(CSF1R):c.2294G>A (p.Gly765Asp)	CSF1R	5	150057312	Hereditary diffuse leukoencephalopathy with spheroids
NM_000506.3(F2):c.940C>T (p.Arg314Cys)	F2	11	46726563	Hereditary factor II deficiency disease
NM_000506.3(F2):c.1381C>T (p.Arg461Trp)	F2	11	46728746	Hereditary factor II deficiency disease
NM_000506.3(F2):c.1273C>T (p.Arg425Cys)	F2	11	46728138	Hereditary factor II deficiency disease
NM_000506.3(F2):c.1027G>A (p.Glu343Lys)	F2	11	46726734	Hereditary factor II deficiency disease
NM_000506.3(F2):c.1054G>A (p.Glu352Lys)	F2	11	46726761	Hereditary factor II deficiency disease
NM_000506.4(F2):c.1274G>A (p.Arg425His)	F2	11	46728139	Hereditary factor II deficiency disease
NM_000133.3(F9):c.169C>T (p.Gln57Ter)	F9	X	139537090	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.79G>A (p.Glu27Lys)	F9	X	139530843	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.223C>T (p.Arg75Ter)	F9	X	139537144	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.224G>A (p.Arg75Gln)	F9	X	139537145	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.316G>A (p.Gly106Ser)	F9	X	139541114	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.571C>T (p.Arg191Cys)	F9	X	139551112	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.572G>A (p.Arg191His)	F9	X	139551113	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.880C>T (p.Arg294Ter)	F9	X	139561565	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.655C>T (p.Gln219Ter)	F9	X	139551196	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.676C>T (p.Arg226Trp)	F9	X	139551217	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.709C>T (p.Gln237Ter)	F9	X	139551250	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.697G>A (p.Ala233Thr)	F9	X	139551238	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.881G>A (p.Arg294Gln)	F9	X	139561566	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.892C>T (p.Arg298Ter)	F9	X	139561577	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.998C>T (p.Pro333Leu)	F9	X	139561683	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1025C>T (p.Thr342Met)	F9	X	139561710	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1069G>A (p.Gly357Arg)	F9	X	139561754	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1135C>T (p.Arg379Ter)	F9	X	139561820	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1136G>A (p.Arg379Gln)	F9	X	139561821	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1150C>T (p.Arg384Ter)	F9	X	139561835	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1217C>T (p.Ser406Leu)	F9	X	139561902	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1307C>T (p.Ala436Val)	F9	X	139561992	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1324G>A (p.Gly442Arg)	F9	X	139562009	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.484C>T (p.Arg162Ter)	F9	X	139548455	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.328G>A (p.Asp110Asn)	F9	X	139541126	Hereditary factor IX deficiency disease
NM_000133.3(F9):c.1070G>A (p.Gly357Glu)	F9	X	139561755	Hereditary factor IX deficiency disease

NM_000133.3(F9):c.835G>A (p.Ala279Thr)	F9	X	139560852	Hereditary factor IX deficiency disease
NM_000132.3(F8):c.6976C>T (p.Arg2326Ter)	F8	X	154837677	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.6682C>T (p.Arg2228Ter)	F8	X	154861759	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.6403C>T (p.Arg2135Ter)	F8	X	154896103	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.6496C>T (p.Arg2166Ter)	F8	X	154863161	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.601+1632G>A	F8	X	154991304	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.6683G>A (p.Arg2228Gln)	F8	X	154861758	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.5879G>A (p.Arg1960Gln)	F8	X	154904025	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.5113C>T (p.Gln1705Ter)	F8	X	154928677	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.5122C>T (p.Arg1708Cys)	F8	X	154928668	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.5878C>T (p.Arg1960Ter)	F8	X	154904026	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1171C>T (p.Arg391Cys)	F8	X	154966526	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.6977G>A (p.Arg2326Gln)	F8	X	154837676	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.566C>T (p.Ser189Leu)	F8	X	154992971	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1063C>T (p.Arg355Ter)	F8	X	154966634	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1336C>T (p.Arg446Ter)	F8	X	154966077	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.5167G>A (p.Glu1723Lys)	F8	X	154928623	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.6955C>T (p.Pro2319Ser)	F8	X	154837698	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.43C>T (p.Arg15Ter)	F8	X	155022510	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.266G>A (p.Gly89Asp)	F8	X	154997095	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.410C>T (p.Thr137Ile)	F8	X	154993127	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.493C>T (p.Pro165Ser)	F8	X	154993044	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.541G>A (p.Val181Met)	F8	X	154992996	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.797G>A (p.Gly266Glu)	F8	X	154969543	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.822G>A (p.Trp274Ter)	F8	X	154969518	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.832G>A (p.Gly278Arg)	F8	X	154969508	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.881C>T (p.Thr294Ile)	F8	X	154969459	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.923C>T (p.Ser308Leu)	F8	X	154969417	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.986G>A (p.Cys329Tyr)	F8	X	154969354	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1175C>T (p.Ser392Leu)	F8	X	154966522	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1492G>A (p.Gly498Arg)	F8	X	154961120	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1630G>A (p.Asp544Asn)	F8	X	154957079	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1636C>T (p.Arg546Trp)	F8	X	154957073	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1648C>T (p.Arg550Cys)	F8	X	154957061	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1649G>A (p.Arg550His)	F8	X	154957060	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1730C>T (p.Ser577Phe)	F8	X	154956979	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1804C>T (p.Arg602Ter)	F8	X	154953991	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1957G>A (p.Val653Met)	F8	X	154947854	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.1988C>T (p.Ala663Val)	F8	X	154947823	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.2149C>T (p.Arg717Trp)	F8	X	154931641	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.2167G>A (p.Ala723Thr)	F8	X	154931623	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.2215G>A (p.Glu739Lys)	F8	X	154931575	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.3169G>A (p.Glu1057Lys)	F8	X	154930621	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.5123G>A (p.Arg1708His)	F8	X	154928667	Hereditary factor VIII deficiency disease
NM_000132.3(F8):c.5143C>T (p.Arg1715Ter)	F8	X	154928647	Hereditary factor VIII deficiency disease

NM_000132.3(F8):c.5305G>A (p.Gly1769Arg)	F8	X	154906488	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5336G>A (p.Gly1779Glu)	F8	X	154906457	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5399G>A (p.Arg1800His)	F8	X	154904998	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5398C>T (p.Arg1800Cys)	F8	X	154904999	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5422C>T (p.Leu1808Phe)	F8	X	154904975	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5443C>T (p.Gln1815Ter)	F8	X	154904954	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5526G>A (p.Met1842Ile)	F8	X	154904871	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5530C>T (p.Pro1844Ser)	F8	X	154904867	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5558C>T (p.Ala1853Val)	F8	X	154904839	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5593G>A (p.Asp1865Asn)	F8	X	154904518	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5677C>T (p.Gln1893Ter)	F8	X	154904434	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5710G>A (p.Glu1904Lys)	F8	X	154904401	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5882G>A (p.Trp1961Ter)	F8	X	154904022	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5900G>A (p.Gly1967Asp)	F8	X	154904004	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5938C>T (p.His1980Tyr)	F8	X	154903966	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.5953C>T (p.Arg1985Ter)	F8	X	154903951	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6046C>T (p.Arg2016Trp)	F8	X	154902120	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6263C>T (p.Ser2088Phe)	F8	X	154899876	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6506G>A (p.Arg2169His)	F8	X	154863151	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6518C>T (p.Thr2173Ile)	F8	X	154863139	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6532C>T (p.Arg2178Cys)	F8	X	154863125	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6533G>A (p.Arg2178His)	F8	X	154863124	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6545G>A (p.Arg2182His)	F8	X	154863112	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6544C>T (p.Arg2182Cys)	F8	X	154863113	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6865C>T (p.Gln2289Ter)	F8	X	154860467	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6956C>T (p.Pro2319Leu)	F8	X	154837697	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6967C>T (p.Arg2323Cys)	F8	X	154837686	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.6968G>A (p.Arg2323His)	F8	X	154837685	Hereditary factor VIII deficiency disease	
NM_000132.3(F8):c.902G>A (p.Arg301His)	F8	X	154969438	Hereditary factor VIII deficiency disease;not provided	
NM_000132.3(F8):c.1834C>T (p.Arg612Cys)	F8	X	154953961	Hereditary factor VIII deficiency disease;not provided	
NM_000128.3(F11):c.1716+1G>A	F11	4	186287824	Hereditary factor XI deficiency disease	
NM_000128.3(F11):c.976C>T (p.Arg326Cys)	F11	4	186280333	Hereditary factor XI deficiency disease	
NM_000128.3(F11):c.1289C>T (p.Ala430Val)	F11	4	186284245	Hereditary factor XI deficiency disease	
NM_000128.3(F11):c.400C>T (p.Gln134Ter)	F11	4	186274190	Hereditary factor XI deficiency disease	
NM_000128.3(F11):c.1432G>A (p.Gly478Arg)	F11	4	186285765	Hereditary factor XI deficiency disease	
NM_000128.3(F11):c.1288G>A (p.Ala430Thr)	F11	4	186284244	Hereditary factor XI deficiency disease	
NM_000128.3(F11):c.326-1G>A	F11	4	186274115	Hereditary factor XI deficiency disease	
NM_000035.3(ALDOB):c.10C>T (p.Arg4Ter)	ALDOB	9	101430878	Hereditary fructosuria	
NM_000035.3(ALDOB):c.178C>T (p.Arg60Ter)	ALDOB	9	101429901	Hereditary fructosuria	
NM_000035.3(ALDOB):c.324+1G>A	ALDOB	9	101429754	Hereditary fructosuria	
NM_000020.2(ACVRL1):c.430C>T (p.Arg144Ter)	ACVRL1	12	51913675	Hereditary hemorrhagic telangiectasia type 2	
NM_000020.2(ACVRL1):c.822G>A (p.Trp274Ter)	ACVRL1	12	51915274	Hereditary hemorrhagic telangiectasia type 2	
NM_000020.2(ACVRL1):c.1121G>A (p.Arg374Gln)	ACVRL1	12	51916108	Hereditary hemorrhagic telangiectasia type 2	
NM_000020.2(ACVRL1):c.1217G>A (p.Trp406Ter)	ACVRL1	12	51916204	Hereditary hemorrhagic telangiectasia type 2	

NM_000020.2(ACVRL1):c.1232G>A (p.Arg411Gln)	ACVRL1	12	51916219	Hereditary hemorrhagic telangiectasia type 2;Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia
NM_000020.2(ACVRL1):c.1450C>T (p.Arg484Trp)	ACVRL1	12	51920831	Hereditary hemorrhagic telangiectasia type 2;Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia
NM_000020.2(ACVRL1):c.1031G>A (p.Cys344Tyr)	ACVRL1	12	51915483	Hereditary hemorrhagic telangiectasia type 2;Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia
NM_000020.2(ACVRL1):c.1120C>T (p.Arg374Trp)	ACVRL1	12	51916107	Hereditary hemorrhagic telangiectasia type 2;Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia;not provided
NM_000020.2(ACVRL1):c.1231C>T (p.Arg411Trp)	ACVRL1	12	51916218	Hereditary hemorrhagic telangiectasia type 2;Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia;not provided
NM_002529.3(NTRK1):c.25C>T (p.Gln9Ter)	NTRK1	1	156860959	Hereditary insensitivity to pain with anhidrosis
NM_001007792.1(NTRK1):c.1976C>T (p.Pro659Leu)	NTRK1	1	156880036	Hereditary insensitivity to pain with anhidrosis
NM_002529.3(NTRK1):c.1550G>A (p.Gly517Glu)	NTRK1	1	156876128	Hereditary insensitivity to pain with anhidrosis
NM_000143.3(FH):c.905-1G>A	FH	1	241504246	Hereditary leiomyomatosis and renal cell cancer
NM_000143.3(FH):c.133-1G>A	FH	1	241517317	Hereditary leiomyomatosis and renal cell cancer
NM_000143.3(FH):c.322C>T (p.Gln108Ter)	FH	1	241513659	Hereditary leiomyomatosis and renal cell cancer
NM_000143.3(FH):c.1126C>T (p.Gln376Ter)	FH	1	241502553	Hereditary leiomyomatosis and renal cell cancer;not provided
NM_000143.3(FH):c.952C>T (p.His318Tyr)	FH	1	241504198	Hereditary leiomyomatosis and renal cell cancer;not provided
NM_000143.3(FH):c.139C>T (p.Gln47Ter)	FH	1	241517310	Hereditary leiomyomatosis and renal cell cancer;not provided
NM_000143.3(FH):c.1500G>A (p.Trp500Ter)	FH	1	241497861	Hereditary leiomyomatosis and renal cell cancer;not provided
NM_000304.3(PMP22):c.199G>A (p.Ala67Thr)	PMP22	17	15239591	Hereditary liability to pressure palsies
NM_000304.3(PMP22):c.117G>A (p.Trp39Ter)	PMP22	17	15259155	Hereditary liability to pressure palsies
NM_182925.4(FLT4):c.2569G>A (p.Gly857Arg)	FLT4	5	180619743	Hereditary lymphedema type I
NM_182925.4(FLT4):c.3341C>T (p.Pro1114Leu)	FLT4	5	180613101	Hereditary lymphedema type I
NM_182925.4(FLT4):c.2632G>A (p.Val878Met)	FLT4	5	180619680	Hereditary lymphedema type I
NM_182925.4(FLT4):c.3316G>A (p.Glu1106Lys)	FLT4	5	180614083	Hereditary lymphedema type I
NM_182925.4(FLT4):c.2563G>A (p.Ala855Thr)	FLT4	5	180619749	Hereditary lymphedema type I
NM_014874.3(MFN2):c.617C>T (p.Thr206Ile)	MFN2	1	11998787	Hereditary motor and sensory neuropathy with optic atrophy
NM_001376.4(DYNC1H1):c.1792C>T (p.Arg598Cys)	DYNC1H1	14	101986017	Hereditary motor and sensory neuropathy;Spinal muscular atrophy, lower extremity predominant 1, autosomal dominant;not provided
NM_001267550.2(TTN):c.95195C>T (p.Pro31732Leu)	TTN	2	178546041	Hereditary myopathy with early respiratory failure
NM_000179.2(MSH6):c.1444C>T (p.Arg482Ter)	MSH6	2	47799427	Hereditary nonpolyposis colorectal cancer type 5;Lynch syndrome;not provided
NM_003242.5(TGFBR2):c.1583G>A (p.Arg528His)	TGFBR2	3	30691478	Hereditary nonpolyposis colorectal cancer type 6;Loeys-Dietz syndrome;Loeys-Dietz syndrome 2;not provided
NM_002769.4(PRSS1):c.235G>A (p.Glu79Lys)	PRSS1	7	142751808	Hereditary pancreatitis
NM_002769.4(PRSS1):c.364C>T (p.Arg122Cys)	PRSS1	7	142751937	Hereditary pancreatitis
NM_002769.4(PRSS1):c.346C>T (p.Arg116Cys)	PRSS1	7	142751919	Hereditary pancreatitis
NM_002769.4(PRSS1):c.47C>T (p.Ala16Val)	PRSS1	7	142750561	Hereditary pancreatitis
NM_003122.4(SPINK1):c.199C>T (p.Arg67Cys)	SPINK1	5	147824702	Hereditary pancreatitis
NM_007272.2(CTRC):c.217G>A (p.Ala73Thr)	CTRC	1	15440577	Hereditary pancreatitis
NM_002769.4(PRSS1):c.365G>A (p.Arg122His)	PRSS1	7	142751938	Hereditary pancreatitis;not provided
NM_007272.2(CTRC):c.164G>A (p.Trp55Ter)	CTRC	1	15440524	Hereditary pancreatitis;Pancreatitis, chronic, susceptibility to
NM_003002.3(SDHD):c.242C>T (p.Pro81Leu)	SDHD	11	112088939	Hereditary Paraganglioma-Pheochromocytoma Syndromes;Hereditary cancer-predisposing syndrome;Paragangliomas 1;Paragangliomas 1 with sensorineural hearing loss;Pheochromocytoma;not provided

NM_003002.3(SDHD):c.112C>T (p.Arg38Ter)	SDHD	11	112087916	Hereditary Paraganglioma-Pheochromocytoma Syndromes;Paraganglioma and gastric stromal sarcoma;Paragangliomas 1;Pheochromocytoma;not provided
NM_213655.4(WNK1):c.2575C>T (p.Gln859Ter)	WNK1	12	868046	Hereditary sensory and autonomic neuropathy type IIA
NM_001184985.1(WNK1):c.3046C>T (p.Gln1016Ter)	WNK1	12	868772	Hereditary sensory and autonomic neuropathy type IIA
NM_001034850.2(RETREG1):c.433C>T (p.Gln145Ter)	RETREG1	5	16565788	Hereditary sensory and autonomic neuropathy type IIA;Hereditary sensory and autonomic neuropathy type IIB
NM_213655.4(WNK1):c.3226C>T (p.Arg1076Ter)	WNK1	12	868697	Hereditary sensory and autonomic neuropathy type IIA;not provided
NM_000032.4(ALAS2):c.871G>A (p.Gly291Ser)	ALAS2	X	55017618	Hereditary sideroblastic anemia
NM_000032.4(ALAS2):c.514G>A (p.Ala172Thr)	ALAS2	X	55021176	Hereditary sideroblastic anemia
NM_000032.4(ALAS2):c.1231C>T (p.Arg411Cys)	ALAS2	X	55014953	Hereditary sideroblastic anemia
NM_000032.4(ALAS2):c.1184G>A (p.Cys395Tyr)	ALAS2	X	55015000	Hereditary sideroblastic anemia
NM_000032.4(ALAS2):c.475G>A (p.Asp159Asn)	ALAS2	X	55021215	Hereditary sideroblastic anemia
NM_000032.4(ALAS2):c.1354C>T (p.Arg452Cys)	ALAS2	X	55014830	Hereditary sideroblastic anemia;not provided
NM_001244008.1(KIF1A):c.206C>T (p.Ser69Leu)	KIF1A	2	240788208	Hereditary spastic paraplegia
NM_003664.4(AP3B1):c.1525C>T (p.Arg509Ter)	AP3B1	5	78141268	Hermansky Pudlak syndrome 2
NM_003664.4(AP3B1):c.716G>A (p.Trp239Ter)	AP3B1	5	78216125	Hermansky Pudlak syndrome 2
NM_000195.4(HPS1):c.398+5G>A	HPS1	10	98435267	Hermansky-Pudlak syndrome 1
NM_000195.4(HPS1):c.1749G>A (p.Trp583Ter)	HPS1	10	98420153	Hermansky-Pudlak syndrome 1
NM_000195.4(HPS1):c.391C>T (p.Arg131Ter)	HPS1	10	98435279	Hermansky-Pudlak syndrome 1
NM_032122.4(DTNBP1):c.177G>A (p.Trp59Ter)	DTNBP1	6	15637789	Hermansky-Pudlak syndrome 1;Hermansky-Pudlak syndrome 7
NM_032383.4(HPS3):c.1163+1G>A	HPS3	3	149145547	Hermansky-Pudlak syndrome 3
NM_032383.4(HPS3):c.1189C>T (p.Arg397Trp)	HPS3	3	149150624	Hermansky-Pudlak syndrome 3
NM_032383.4(HPS3):c.2888-1612G>A	CP;HPS3	3	149170483	Hermansky-Pudlak syndrome 3
NM_022081.5(HPS4):c.1891C>T (p.Gln631Ter)	HPS4	22	26457923	Hermansky-Pudlak syndrome 4
NM_022081.5(HPS4):c.541C>T (p.Gln181Ter)	HPS4	22	26470774	Hermansky-Pudlak syndrome 4
NM_022081.5(HPS4):c.649C>T (p.Arg217Ter)	HPS4	22	26468571	Hermansky-Pudlak syndrome 4
NM_181507.1(HPS5):c.2593C>T (p.Arg865Ter)	HPS5	11	18287659	Hermansky-Pudlak syndrome 5
NM_024747.5(HPS6):c.1234C>T (p.Gln412Ter)	HPS6	10	102066708	Hermansky-Pudlak syndrome 6
NM_024747.5(HPS6):c.913C>T (p.Gln305Ter)	HPS6	10	102066387	Hermansky-Pudlak syndrome 6
NM_024747.5(HPS6):c.815C>T (p.Thr272Ile)	HPS6	10	102066289	Hermansky-Pudlak syndrome 6
NM_024747.5(HPS6):c.223C>T (p.Gln75Ter)	HPS6	10	102065697	Hermansky-Pudlak syndrome 6
NM_032122.4(DTNBP1):c.307C>T (p.Gln103Ter)	DTNBP1	6	15627391	Hermansky-Pudlak syndrome 7
NM_012388.3(BLOC1S6):c.232C>T (p.Gln78Ter)	BLOC1S6	15	45603107	Hermansky-Pudlak syndrome 9
NM_003265.2(TLR3):c.1660C>T (p.Pro554Ser)	TLR3	4	186083346	Herpes simplex encephalitis 2
NM_145725.2(TRAF3):c.352C>T (p.Arg118Trp)	TRAF3	14	102875678	Herpes simplex encephalitis, susceptibility to, 3
NM_182919.3(TICAM1):c.421C>T (p.Arg141Ter)	TICAM1	19	4817957	Herpes simplex encephalitis, susceptibility to, 4
NM_182919.3(TICAM1):c.557C>T (p.Ser186Leu)	TICAM1	19	4817821	Herpes simplex encephalitis, susceptibility to, 4
NM_032545.3(CFC1):c.334C>T (p.Arg112Cys)	CFC1	2	130597896	Heterotaxy, visceral, 2, autosomal
NM_001106.3(ACVR2B):c.1480G>A (p.Val494Ile)	ACVR2B	3	38483273	Heterotaxy, visceral, 4, autosomal
NM_145020.4(CFAP53):c.121C>T (p.Arg41Ter)	CFAP53	18	50262168	Heterotaxy, visceral, 6, autosomal
NM_003413.3(ZIC3):c.968C>T (p.Thr323Met)	ZIC3	X	137567659	Heterotaxy, visceral, X-linked
NM_003413.3(ZIC3):c.745C>T (p.Gln249Ter)	ZIC3	X	137567436	Heterotaxy, visceral, X-linked
NM_178151.2(DCX):c.305G>A (p.Arg102His)	DCX	X	111410094	Heterotopia
NM_178151.2(DCX):c.907C>T (p.Arg303Ter)	DCX	X	111330943	Heterotopia
NM_178151.2(DCX):c.809-1G>A	DCX	X	111331042	Heterotopia

NM_178151.2(DCX):c.667G>A (p.Gly223Arg)	DCX	X	111401028	Heterotopia
NM_178151.2(DCX):c.557G>A (p.Arg186His)	DCX	X	111401138	Heterotopia
NM_178151.2(DCX):c.556C>T (p.Arg186Cys)	DCX	X	111401139	Heterotopia
NM_178151.2(DCX):c.532C>T (p.Arg178Cys)	DCX	X	111401163	Heterotopia
NM_178151.2(DCX):c.478C>T (p.Gln160Ter)	DCX	X	111401217	Heterotopia
NM_178151.2(DCX):c.386C>T (p.Ser129Leu)	DCX	X	111401309	Heterotopia
NM_178151.2(DCX):c.364G>A (p.Gly122Arg)	DCX	X	111410035	Heterotopia
NM_178151.2(DCX):c.304C>T (p.Arg102Cys)	DCX	X	111410095	Heterotopia
NM_178151.2(DCX):c.301G>A (p.Val101Met)	DCX	X	111410098	Heterotopia
NM_178151.2(DCX):c.265C>T (p.Arg89Ter)	DCX	X	111410134	Heterotopia
NM_178151.2(DCX):c.232C>T (p.Arg78Cys)	DCX	X	111410167	Heterotopia
NM_178151.2(DCX):c.218C>T (p.Ser73Phe)	DCX	X	111410181	Heterotopia
NM_178151.2(DCX):c.182G>A (p.Gly61Glu)	DCX	X	111410217	Heterotopia
NM_178151.2(DCX):c.176G>A (p.Arg59His)	DCX	X	111410223	Heterotopia
NM_178151.2(DCX):c.130C>T (p.Gln44Ter)	DCX	X	111410269	Heterotopia
NM_178151.2(DCX):c.115C>T (p.Arg39Ter)	DCX	X	111410284	Heterotopia
NM_178151.2(DCX):c.95G>A (p.Ser32Asn)	DCX	X	111410304	Heterotopia
NM_178151.2(DCX):c.55C>T (p.Arg19Ter)	DCX	X	111410344	Heterotopia
NM_006420.2(ARFGEF2):c.1958+1G>A	ARFGEF2	20	48976200	Heterotopia, periventricular, autosomal recessive
NM_178151.2(DCX):c.781C>T (p.Gln261Ter)	DCX	X	111333078	Heterotopia;Lissencephaly, X-linked
NM_178151.2(DCX):c.587G>A (p.Arg196His)	DCX	X	111401108	Heterotopia;Lissencephaly, X-linked;Subcortical laminar heterotopia, X-linked
NM_178151.2(DCX):c.574C>T (p.Arg192Trp)	DCX	X	111401121	Heterotopia;Lissencephaly, X-linked;Subcortical laminar heterotopia, X-linked;not provided
NM_178151.2(DCX):c.814C>T (p.Arg272Ter)	DCX	X	111331036	Heterotopia;not provided
NM_178151.2(DCX):c.586C>T (p.Arg196Cys)	DCX	X	111401109	Heterotopia;not provided
NM_006783.4(GJB6):c.263C>T (p.Ala88Val)	GJB6	13	20223218	Hidrotic ectodermal dysplasia syndrome
NM_006783.4(GJB6):c.31G>A (p.Gly11Arg)	GJB6	13	20223450	Hidrotic ectodermal dysplasia syndrome;Nonsyndromic hearing loss and deafness;not provided
NM_020975.4(RET):c.2690G>A (p.Arg897Gln)	RET	10	43120163	Hirschsprung disease 1
NM_020975.4(RET):c.95C>T (p.Ser32Leu)	RET	10	43100480	Hirschsprung disease 1
NM_020975.4(RET):c.191C>T (p.Pro64Leu)	RET	10	43100576	Hirschsprung disease 1
NM_020975.4(RET):c.538C>T (p.Arg180Ter)	RET	10	43102542	Hirschsprung disease 1
NM_020975.4(RET):c.989G>A (p.Arg330Gln)	RET	10	43106497	Hirschsprung disease 1
NM_020975.4(RET):c.938G>A (p.Arg313Gln)	RET	10	43106446	Hirschsprung disease 1
NM_020975.4(RET):c.692G>A (p.Arg231His)	RET	10	43105018	Hirschsprung disease 1
NM_000115.4(EDNRB):c.824G>A (p.Trp275Ter)	EDNRB	13	77901185	Hirschsprung disease 2
NM_001113348.1(ECE1):c.2212C>T (p.Arg738Cys)	ECE1	1	21220008	Hirschsprung disease, cardiac defects, and autonomic dysfunction
NM_000536.3(RAG2):c.283G>A (p.Gly95Arg)	RAG2	11	36593886	Histiocytic medullary reticulosis
NM_000448.2(RAG1):c.1682G>A (p.Arg561His)	RAG1	11	36574986	Histiocytic medullary reticulosis
NM_000448.2(RAG1):c.1187G>A (p.Arg396His)	RAG1	11	36574491	Histiocytic medullary reticulosis
NM_000448.2(RAG1):c.983G>A (p.Cys328Tyr)	RAG1	11	36574287	Histiocytic medullary reticulosis
NM_000448.2(RAG1):c.1186C>T (p.Arg396Cys)	RAG1	11	36574490	Histiocytic medullary reticulosis;not provided
NM_000536.3(RAG2):c.686G>A (p.Arg229Gln)	RAG2	11	36593483	Histiocytic medullary reticulosis;Severe combined immunodeficiency, B cell-negative
NM_000448.2(RAG1):c.1681C>T (p.Arg561Cys)	RAG1	11	36574985	Histiocytic medullary reticulosis;Severe combined immunodeficiency, B cell-negative
NM_018344.5(SLC29A3):c.1279G>A (p.Gly427Ser)	SLC29A3	10	71362459	Histiocytosis-lymphadenopathy plus syndrome
NM_018344.5(SLC29A3):c.1088G>A (p.Arg363Gln)	SLC29A3	10	71362268	Histiocytosis-lymphadenopathy plus syndrome
NM_018344.5(SLC29A3):c.1087C>T (p.Arg363Trp)	SLC29A3	10	71362267	Histiocytosis-lymphadenopathy plus syndrome

NM_018344.5(SLC29A3):c.1157G>A (p.Arg386Gln)	SLC29A3	10	71362337	Histiocytosis-lymphadenopathy plus syndrome
NM_018344.5(SLC29A3):c.1228C>T (p.Gln410Ter)	SLC29A3	10	71362408	Histiocytosis-lymphadenopathy plus syndrome
NM_018344.5(SLC29A3):c.300+1G>A	SLC29A3	10	71323055	Histiocytosis-lymphadenopathy plus syndrome
NM_018344.5(SLC29A3):c.73C>T (p.Arg25Ter)	SLC29A3	10	71322827	Histiocytosis-lymphadenopathy plus syndrome
NM_018344.5(SLC29A3):c.1309G>A (p.Gly437Arg)	SLC29A3	10	71362489	Histiocytosis-lymphadenopathy plus syndrome;not provided
NM_000034.3(ALDOA):c.619G>A (p.Glu207Lys)	ALDOA	16	30069384	HNSHA due to aldolase A deficiency
NM_173546.2(KLHDC8B):c.-158C>T	KLHDC8B	3	49171662	Hodgkin lymphoma
NM_000411.6(HLCS):c.1711G>A (p.Asp571Asn)	HLCS	21	36759811	Holocarboxylase synthetase deficiency
NM_000411.6(HLCS):c.1522C>T (p.Arg508Trp)	HLCS	21	36765170	Holocarboxylase synthetase deficiency
NM_000411.6(HLCS):c.1741G>A (p.Gly581Ser)	HLCS	21	36759781	Holocarboxylase synthetase deficiency
NM_000411.6(HLCS):c.1519+5G>A	HLCS	21	36767213	Holocarboxylase synthetase deficiency
NM_000411.6(HLCS):c.1624C>T (p.Gln542Ter)	HLCS	21	36765068	Holocarboxylase synthetase deficiency
NM_000411.6(HLCS):c.1648G>A (p.Val550Met)	HLCS	21	36765044	Holocarboxylase synthetase deficiency;not provided
NM_005413.3(SIX3):c.206G>A (p.Gly69Asp)	SIX3	2	44942310	Holoprosencephaly 2
NM_000193.3(SHH):c.91G>A (p.Gly31Arg)	SHH	7	155812032	Holoprosencephaly 3
NM_000193.3(SHH):c.298C>T (p.Gln100Ter)	SHH	7	155811825	Holoprosencephaly 3
NM_000193.3(SHH):c.676G>A (p.Ala226Thr)	SHH	7	155803613	Holoprosencephaly 3
NM_000193.3(SHH):c.1147G>A (p.Ala383Thr)	SHH	7	155803142	Holoprosencephaly 3
NM_000193.3(SHH):c.625C>T (p.Gln209Ter)	SHH	7	155803664	Holoprosencephaly 3
NM_000193.3(SHH):c.664G>A (p.Asp222Asn)	SHH	7	155803625	Holoprosencephaly 3
NM_000193.3(SHH):c.383G>A (p.Trp128Ter)	SHH	7	155806475	Holoprosencephaly 3;Single upper central incisor;not provided
NM_170695.3(TGIF1):c.872C>T (p.Ser291Phe)	TGIF1	18	3457606	Holoprosencephaly 4
NM_007129.4(ZIC2):c.793C>T (p.Gln265Ter)	ZIC2	13	99982857	Holoprosencephaly 5
NM_000264.3(PTCH1):c.1177G>A (p.Ala393Thr)	PTCH1	9	95479038	Holoprosencephaly 7
NM_005270.4(GLI2):c.1323G>A (p.Trp441Ter)	GLI2	2	120975064	Holoprosencephaly 9
NM_005270.4(GLI2):c.1885G>A (p.Glu629Lys)	GLI2	2	120984672	Holoprosencephaly 9
NM_000192.3(TBX5):c.238G>A (p.Gly80Arg)	TBX5	12	114401830	Holt-Oram syndrome
NM_005957.4(MTHFR):c.1743G>A (p.Met581Ile)	MTHFR	1	11791216	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.1753-18G>A	MTHFR	1	11790916	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.1683G>A (p.Trp561Ter)	MTHFR	1	11791276	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.1320G>A (p.Ser440=)	MTHFR	1	11794385	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.1088G>A (p.Arg363His)	MTHFR	1	11794807	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.1060C>T (p.His354Tyr)	MTHFR	1	11794835	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.1042C>T (p.Pro348Ser)	MTHFR	1	11794853	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.760C>T (p.Pro254Ser)	MTHFR	1	11796226	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.587G>A (p.Gly196Asp)	MTHFR	1	11796399	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.548G>A (p.Arg183Gln)	MTHFR	1	11800250	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.379C>T (p.His127Tyr)	MTHFR	1	11801257	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.337G>A (p.Ala113Thr)	MTHFR	1	11801299	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.244C>T (p.Arg82Trp)	MTHFR	1	11801392	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.137G>A (p.Arg46Gln)	MTHFR	1	11802980	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.136C>T (p.Arg46Trp)	MTHFR	1	11802981	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.1033C>T (p.Arg345Cys)	MTHFR	1	11794862	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.416C>T (p.Thr139Met)	MTHFR	1	11801220	Homocysteinemia due to MTHFR deficiency
NM_005957.4(MTHFR):c.236+1G>A	MTHFR	1	11802880	Homocysteinemia due to MTHFR deficiency

NM_000071.2(CBS):c.374G>A (p.Arg125Gln)	CBS	21	43066320	Homocystinuria due to CBS deficiency
NM_000071.2(CBS):c.954+1G>A	CBS	21	43062952	Homocystinuria due to CBS deficiency
NM_000071.2(CBS):c.1058C>T (p.Thr353Met)	CBS	21	43060528	Homocystinuria due to CBS deficiency;Homocystinuria, pyridoxine-nonresponsive;Thoracic aortic aneurysm and aortic dissection;not provided
NM_000071.2(CBS):c.430G>A (p.Glu144Lys)	CBS	21	43066264	Homocystinuria due to CBS deficiency;Homocystinuria, pyridoxine-responsive
NM_000071.2(CBS):c.797G>A (p.Arg266Lys)	CBS	21	43063931	Homocystinuria due to CBS deficiency;Homocystinuria, pyridoxine-responsive
NM_000071.2(CBS):c.1330G>A (p.Asp444Asn)	CBS	21	43058862	Homocystinuria due to CBS deficiency;Homocystinuria, pyridoxine-responsive;not provided
NM_000071.2(CBS):c.1006C>T (p.Arg336Cys)	CBS	21	43062344	Homocystinuria due to CBS deficiency;not provided
NM_000071.2(CBS):c.1136G>A (p.Arg379Gln)	CBS	21	43060450	Homocystinuria due to CBS deficiency;not provided
NM_000071.2(CBS):c.1039G>A (p.Gly347Ser)	CBS	21	43062311	Homocystinuria due to CBS deficiency;not provided
NM_000071.2(CBS):c.770C>T (p.Thr257Met)	CBS	21	43063958	Homocystinuria due to CBS deficiency;not provided
NM_000071.2(CBS):c.785C>T (p.Thr262Met)	CBS	21	43063943	Homocystinuria due to CBS deficiency;not provided
NM_000071.2(CBS):c.1111G>A (p.Val371Met)	CBS	21	43060475	Homocystinuria due to CBS deficiency;not provided
NM_000071.2(CBS):c.969G>A (p.Trp323Ter)	CBS	21	43062381	Homocystinuria due to CBS deficiency;not provided
NM_000071.2(CBS):c.146C>T (p.Pro49Leu)	CBS	21	43072048	Homocystinuria due to CBS deficiency;Thoracic aortic aneurysm and aortic dissection;not provided
NM_005957.4(MTHFR):c.547C>T (p.Arg183Ter)	MTHFR	1	11800251	Homocystinuria due to MTHFR deficiency
NM_005957.4(MTHFR):c.470G>A (p.Arg157Gln)	MTHFR	1	11801166	Homocystinuria due to MTHFR deficiency
NM_005957.4(MTHFR):c.1129C>T (p.Arg377Cys)	MTHFR	1	11794766	Homocystinuria due to MTHFR deficiency
NM_000071.2(CBS):c.572C>T (p.Thr191Met)	CBS	21	43065481	Homocystinuria, pyridoxine-nonresponsive;not provided
NM_000071.2(CBS):c.434C>T (p.Pro145Leu)	CBS	21	43066260	Homocystinuria, pyridoxine-responsive
NM_000071.2(CBS):c.415G>A (p.Gly139Arg)	CBS	21	43066279	Homocystinuria, pyridoxine-responsive
NM_000071.2(CBS):c.341C>T (p.Ala114Val)	CBS	21	43066353	Homocystinuria, pyridoxine-responsive;not provided
NM_000071.2(CBS):c.919G>A (p.Gly307Ser)	CBS	21	43062988	Homocystinuria;Homocystinuria due to CBS deficiency;Homocystinuria, pyridoxine-nonresponsive;not provided
NM_002454.2(MTRR):c.1361C>T (p.Ser454Leu)	MTRR	5	7891405	Homocystinuria-Megaloblastic anemia due to defect in cobalamin metabolism, cblE complementation type
NM_002454.2(MTRR):c.1459G>A (p.Gly487Arg)	MTRR	5	7892815	Homocystinuria-Megaloblastic anemia due to defect in cobalamin metabolism, cblE complementation type;Inborn genetic diseases
NM_024599.5(RHBDF2):c.566C>T (p.Pro189Leu)	RHBDF2	17	76478999	Howel-Evans syndrome
NM_000579.3(CCR5):c.-229C>T	CCR5	3	46370771	Human immunodeficiency virus type 1, increased perinatal transmission of
NM_001123041.2(CCR2):c.190G>A (p.Val64Ile)	CCR2	3	46357717	Human immunodeficiency virus type 1, susceptibility to
NM_000609.6(CXCL12):c.266+535G>A	CXCL12	10	44378102	Human immunodeficiency virus type 1, susceptibility to
NM_000203.4(IDUA):c.1861C>T (p.Arg621Ter)	IDUA	4	1004292	Hurler syndrome
NM_000203.4(IDUA):c.928C>T (p.Gln310Ter)	IDUA	4	1002117	Hurler syndrome
NM_000203.4(IDUA):c.972+1G>A	IDUA	4	1002162	Hurler syndrome
NM_000203.4(IDUA):c.1855C>T (p.Arg619Ter)	IDUA	4	1004286	Hurler syndrome
NM_000203.4(IDUA):c.152G>A (p.Gly51Asp)	IDUA	4	987236	Hurler syndrome;Mucopolysaccharidosis type 1
NM_000203.4(IDUA):c.1205G>A (p.Trp402Ter)	IDUA	4	1002747	Hurler syndrome;Mucopolysaccharidosis type 1;Mucopolysaccharidosis type I;Mucopolysaccharidosis, MPS-I-H/S;Mucopolysaccharidosis, MPS-I-S;not provided
NM_000203.4(IDUA):c.208C>T (p.Gln70Ter)	IDUA	4	987858	Hurler syndrome;Mucopolysaccharidosis type 1;Mucopolysaccharidosis type I;Mucopolysaccharidosis, MPS-I-H/S;Mucopolysaccharidosis, MPS-I-S;not provided
NM_000203.4(IDUA):c.266G>A (p.Arg89Gln)	IDUA	4	987916	Hurler syndrome;Mucopolysaccharidosis type 1;Mucopolysaccharidosis, MPS-I-H/S
NM_000203.4(IDUA):c.1045G>A (p.Asp349Asn)	IDUA	4	1002341	Hurler syndrome;not provided
NM_000203.4(IDUA):c.1650+5G>A	IDUA	4	1003475	Hurler syndrome;not provided
NM_170707.3(LMNA):c.1968G>A (p.Gln656=)	LMNA	1	156138757	Hutchinson-Gilford progeria syndrome, atypical;Hutchinson-Gilford syndrome

NM_170707.3(LMNA):c.1968+5G>A	LMNA	1	156138762	Hutchinson-Gilford progeria syndrome, atypical;Hutchinson-Gilford syndrome
NM_170707.3(LMNA):c.433G>A (p.Glu145Lys)	LMNA	1	156130693	Hutchinson-Gilford progeria syndrome, atypical;Hutchinson-Gilford syndrome;not provided
NM_170707.3(LMNA):c.667G>A (p.Glu223Lys)	LMNA	1	156134832	Hutchinson-Gilford syndrome
NM_170707.3(LMNA):c.1824C>T (p.Gly608=)	LMNA	1	156138613	Hutchinson-Gilford syndrome;Laminopathy;Lethal tight skin contracture syndrome;Mandibuloacral dysostosis;not provided
NM_170707.3(LMNA):c.1968+1G>A	LMNA	1	156138758	Hutchinson-Gilford syndrome;Lethal tight skin contracture syndrome;not provided
NM_170707.3(LMNA):c.1822G>A (p.Gly608Ser)	LMNA	1	156138611	Hutchinson-Gilford syndrome;not provided
NM_170707.3(LMNA):c.1821G>A (p.Val607=)	LMNA	1	156138610	Hutchinson-Gilford syndrome;not provided
NM_058172.5(ANTXR2):c.1179G>A (p.Glu393=)	ANTXR2	4	79983878	Hyaline fibromatosis syndrome
NM_058172.5(ANTXR2):c.314G>A (p.Gly105Asp)	ANTXR2	4	80055996	Hyaline fibromatosis syndrome
NM_001127255.1(NLRP7):c.352+1G>A	NLRP7	19	54940930	Hydatidiform mole
NM_001127255.1(NLRP7):c.2471+1G>A	NLRP7	19	54934488	Hydatidiform mole
NM_001127255.1(NLRP7):c.2077C>T (p.Arg693Trp)	NLRP7	19	54938096	Hydatidiform mole
NM_001127255.1(NLRP7):c.1294C>T (p.Arg432Ter)	NLRP7	19	54939525	Hydatidiform mole
NM_001127255.1(NLRP7):c.2078G>A (p.Arg693Gln)	NLRP7	19	54938095	Hydatidiform mole
NM_001127255.1(NLRP7):c.1951C>T (p.Pro651Ser)	NLRP7	19	54938222	Hydatidiform mole
NM_001080414.3(CCDC88C):c.5058+1G>A	CCDC88C	14	91277921	Hydrocephalus
NM_001080414.3(CCDC88C):c.934C>T (p.Arg312Ter)	CCDC88C	14	91338121	Hydrocephalus
NM_000425.4(L1CAM):c.2278C>T (p.Arg760Ter)	L1CAM	X	153866802	Hydrocephalus due to aqueductal stenosis
NM_001261406.1(MPDZ):c.628C>T (p.Gln210Ter)	MPDZ	9	13222352	Hydrocephalus, nonsyndromic, autosomal recessive 2
NM_003829.4(MPDZ):c.755G>A (p.Trp252Ter)	MPDZ	9	13221493	Hydrocephalus, nonsyndromic, autosomal recessive 2
NM_003829.4(MPDZ):c.5302C>T (p.Gln1768Ter)	MPDZ	9	13119579	Hydrocephalus, nonsyndromic, autosomal recessive 2
NM_000425.4(L1CAM):c.2974C>T (p.Gln992Ter)	L1CAM	X	153864893	Hydrocephalus, X-linked, with congenital idiopathic intestinal pseudoobstruction;not provided
NM_004444.4(EPHB4):c.2216G>A (p.Arg739Gln)	EPHB4	7	100807483	HYDROPS FETALIS, NONIMMUNE, AND/OR ATRIAL SEPTAL DEFECT, SUSCEPTIBILITY TO
NM_015340.3(LARS2):c.1289C>T (p.Ala430Val)	LARS2	3	45491566	Hydrops, lactic acidosis, and sideroblastic anemia
NM_153006.2(NAGS):c.971G>A (p.Trp324Ter)	NAGS	17	44006584	Hyperammonemia, type III
NM_153006.2(NAGS):c.835G>A (p.Ala279Thr)	NAGS	17	44006157	Hyperammonemia, type III
NM_001253823.1(BLVRA):c.52C>T (p.Arg18Ter)	BLVRA	7	43787943	Hyperbiliverdinemia
NM_003052.4(SLC34A1):c.644+1G>A	SLC34A1	5	177387874	Hypercalcemia, infantile, 2
NM_003052.4(SLC34A1):c.1006+1G>A	SLC34A1	5	177393764	Hypercalcemia, infantile, 2;not provided
NM_017429.2(BCO1):c.509C>T (p.Thr170Met)	BCO1	16	81264677	Hypercarotenemia and vitamin a deficiency, autosomal dominant
NM_001218.4(CA12):c.427G>A (p.Glu143Lys)	CA12	15	63345479	Hyperchlorhidrosis, isolated
NM_001218.4(CA12):c.908-1G>A	CA12	15	63327234	Hyperchlorhidrosis, isolated
NM_000041.3(APOE):c.61G>A (p.Glu21Lys)	APOE	19	44907777	Hypercholesterolemia and hypertriglyceridemia, type III
NM_174936.3(PCSK9):c.644G>A (p.Arg215His)	PCSK9	1	55052398	Hypercholesterolemia, autosomal dominant, 3;not provided
NM_015627.2(LDLRAP1):c.65G>A (p.Trp22Ter)	LDLRAP1	1	25543763	Hypercholesterolemia, autosomal recessive
NM_015627.2(LDLRAP1):c.406C>T (p.Gln136Ter)	LDLRAP1	1	25557214	Hypercholesterolemia, autosomal recessive
NM_000824.4(GLRB):c.752G>A (p.Gly251Asp)	GLRB	4	157143807	Hyperekplexia 2
NM_000824.4(GLRB):c.610+5G>A	GLRB	4	157136891	Hyperekplexia 2
NM_004211.4(SLC6A5):c.1888C>T (p.Gln630Ter)	SLC6A5	11	20638477	Hyperekplexia 3
NM_004211.4(SLC6A5):c.1274C>T (p.Thr425Met)	SLC6A5	11	20626721	Hyperekplexia 3
NM_001146040.1(GLRA1):c.896G>A (p.Arg299Gln)	GLRA1	5	151851406	Hyperekplexia hereditary
NM_001146040.1(GLRA1):c.862G>A (p.Val288Met)	GLRA1	5	151851440	Hyperekplexia hereditary
NM_001146040.1(GLRA1):c.884G>A (p.Ser295Asn)	GLRA1	5	151851418	Hyperekplexia hereditary
NM_001146040.1(GLRA1):c.299G>A (p.Arg100His)	GLRA1	5	151859962	Hyperekplexia hereditary

NM_001146040.1(GLRA1):c.737G>A (p.Arg246Gln)	GLRA1	5	151851565	Hyperekplexia hereditary
NM_000171.3(GLRA1):c.1259G>A (p.Arg420His)	GLRA1	5	151822764	Hyperekplexia hereditary;not provided
NM_006206.5(PDGFRA):c.2021C>T (p.Thr674Ile)	PDGFRA	4	54278380	Hypereosinophilic syndrome, idiopathic, resistant to imatinib
NM_000146.3(FTL):c.-168G>A	FTL	19	48965340	Hyperferritinemia cataract syndrome
NM_000146.3(FTL):c.-161C>T	FTL	19	48965347	Hyperferritinemia cataract syndrome
NM_000146.3(FTL):c.89C>T (p.Thr30Ile)	FTL	19	48965596	Hyperferritinemia cataract syndrome
NM_000146.3(FTL):c.-164C>T	FTL	19	48965344	Hyperferritinemia cataract syndrome
NM_000431.3(MVK):c.494C>T (p.Pro165Leu)	MVK	12	109581517	Hyperimmunoglobulin D with periodic fever
NM_000431.3(MVK):c.564G>A (p.Trp188Ter)	MVK	12	109586058	Hyperimmunoglobulin D with periodic fever
NM_000431.3(MVK):c.1000G>A (p.Ala334Thr)	MVK	12	109595142	Hyperimmunoglobulin D with periodic fever;Mevalonic aciduria
NM_000431.3(MVK):c.1129G>A (p.Val377Ile)	MVK	12	109596515	Hyperimmunoglobulin D with periodic fever;Mevalonic aciduria;not provided
NM_000431.3(MVK):c.928G>A (p.Val310Met)	MVK	12	109595070	Hyperimmunoglobulin D with periodic fever;Mevalonic aciduria;not provided
NM_000431.3(MVK):c.604G>A (p.Gly202Arg)	MVK	12	109586098	Hyperimmunoglobulin D with periodic fever;Mevalonic aciduria;Porokeratosis, disseminated superficial actinic 1
NM_000431.3(MVK):c.500C>T (p.Pro167Leu)	MVK	12	109581523	Hyperimmunoglobulin D with periodic fever;not provided
NM_000431.3(MVK):c.1162C>T (p.Arg388Ter)	MVK	12	109596548	Hyperimmunoglobulin D with periodic fever;not provided
NM_000431.3(MVK):c.442G>A (p.Ala148Thr)	MVK	12	109581465	Hyperimmunoglobulin D with periodic fever;not provided
NM_139276.2(STAT3):c.1145G>A (p.Arg382Gln)	STAT3	17	42329642	Hyperimmunoglobulin E syndrome
NM_139276.2(STAT3):c.1268G>A (p.Arg423Gln)	STAT3	17	42329423	Hyperimmunoglobulin E syndrome
NM_139276.2(STAT3):c.1166C>T (p.Thr389Ile)	STAT3	17	42329621	Hyperimmunoglobulin E syndrome
NM_139276.2(STAT3):c.1144C>T (p.Arg382Trp)	STAT3	17	42329643	Hyperimmunoglobulin E syndrome;not provided
NM_139276.2(STAT3):c.1909G>A (p.Val637Met)	STAT3	17	42322474	Hyperimmunoglobulin E syndrome;not provided
NM_000162.3(GCK):c.1363G>A (p.Val455Met)	GCK	7	44145171	Hyperinsulinemic hypoglycemia familial 3
NM_000162.3(GCK):c.1367C>T (p.Ala456Val)	GCK	7	44145167	Hyperinsulinemic hypoglycemia familial 3
NM_000208.3(INSR):c.3602G>A (p.Arg1201Gln)	INSR	19	7120677	Hyperinsulinemic hypoglycemia familial 5;Insulin-resistant diabetes mellitus AND acanthosis nigricans
NM_005327.4(HADH):c.773C>T (p.Pro258Leu)	HADH	4	108033239	Hyperinsulinemic hypoglycemia, familial, 4
NM_005327.4(HADH):c.706C>T (p.Arg236Ter)	HADH	4	108027757	Hyperinsulinemic hypoglycemia, familial, 4
NM_005271.4(GLUD1):c.1519C>T (p.His507Tyr)	GLUD1	10	87053380	Hyperinsulinism-hyperammonemia syndrome
NM_005271.4(GLUD1):c.1495G>A (p.Gly499Ser)	GLUD1	10	87053404	Hyperinsulinism-hyperammonemia syndrome
NM_005271.4(GLUD1):c.1496G>A (p.Gly499Asp)	GLUD1	10	87053403	Hyperinsulinism-hyperammonemia syndrome
NM_005271.4(GLUD1):c.953G>A (p.Arg318Lys)	GLUD1	10	87061021	Hyperinsulinism-hyperammonemia syndrome
NM_005271.4(GLUD1):c.820C>T (p.Arg274Cys)	GLUD1	10	87062757	Hyperinsulinism-hyperammonemia syndrome
NM_005271.4(GLUD1):c.965G>A (p.Arg322His)	GLUD1	10	87061009	Hyperinsulinism-hyperammonemia syndrome
NM_005271.4(GLUD1):c.1493C>T (p.Ser498Leu)	GLUD1	10	87057692	Hyperinsulinism-hyperammonemia syndrome;not provided
NM_000334.4(SCN4A):c.2014C>T (p.Arg672Cys)	SCN4A	17	63959270	Hyperkalemic Periodic Paralysis Type 1;Hypokalemic periodic paralysis 1
NM_000334.4(SCN4A):c.2006G>A (p.Arg669His)	SCN4A	17	63959278	Hyperkalemic Periodic Paralysis Type 1;Hypokalemic periodic paralysis 1;Hypokalemic periodic paralysis, type 2
NM_000334.4(SCN4A):c.2015G>A (p.Arg672His)	SCN4A	17	63959269	Hyperkalemic Periodic Paralysis Type 1;Hypokalemic periodic paralysis 1;Hypokalemic periodic paralysis, type 2
NM_000334.4(SCN4A):c.2023C>T (p.Arg675Trp)	SCN4A	17	63957515	Hyperkalemic Periodic Paralysis Type 1;Normokalemic periodic paralysis, potassium-sensitive
NM_000334.4(SCN4A):c.2024G>A (p.Arg675Gln)	SCN4A	17	63957514	Hyperkalemic Periodic Paralysis Type 1;Normokalemic periodic paralysis, potassium-sensitive
NM_000334.4(SCN4A):c.4343G>A (p.Arg1448His)	SCN4A	17	63941939	Hyperkalemic Periodic Paralysis Type 1;Paramyotonia congenita of von Eulenburg
NM_000334.4(SCN4A):c.4342C>T (p.Arg1448Cys)	SCN4A	17	63941940	Hyperkalemic Periodic Paralysis Type 1;Paramyotonia congenita of von Eulenburg;not provided
NM_000334.4(SCN4A):c.3466G>A (p.Ala1156Thr)	SCN4A	17	63945614	Hyperkalemic Periodic Paralysis Type 1;Paramyotonia congenita/hyperkalemic periodic paralysis

NM_000237.2(LPL):c.607G>A (p.Ala203Thr)	LPL	8	19954185	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.644G>A (p.Gly215Glu)	LPL	8	19954222	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.397C>T (p.Gln133Ter)	LPL	8	19951916	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.701C>T (p.Pro234Leu)	LPL	8	19954279	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.809G>A (p.Arg270His)	LPL	8	19955874	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.506G>A (p.Gly169Glu)	LPL	8	19953386	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.1227G>A (p.Trp409Ter)	LPL	8	19960988	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.829G>A (p.Asp277Asn)	LPL	8	19955894	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.272G>A (p.Trp91Ter)	LPL	8	19951791	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.1081G>A (p.Ala361Thr)	LPL	8	19959322	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.665G>A (p.Gly222Glu)	LPL	8	19954243	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.808C>T (p.Arg270Cys)	LPL	8	19955873	Hyperlipoproteinemia, type I
NM_000237.2(LPL):c.1334G>A (p.Cys445Tyr)	LPL	8	19962126	Hyperlipoproteinemia, type I
NM_178172.5(GPIHBP1):c.194G>A (p.Cys65Tyr)	GPIHBP1	8	143215025	Hyperlipoproteinemia, type ID
NM_005763.3(AASS):c.194G>A (p.Arg65Gln)	AASS	7	122133533	Hyperlysinemia
NM_015359.4(SLC39A14):c.1147G>A (p.Gly383Arg)	SLC39A14	8	22416280	Hypermanganesemia with dystonia 2
NM_018713.2(SLC30A10):c.922C>T (p.Gln308Ter)	SLC30A10	1	219918291	Hypermanganesemia with dystonia, polycythemia and cirrhosis
NM_018713.2(SLC30A10):c.1006C>T (p.His336Tyr)	SLC30A10	1	219915901	Hypermanganesemia with dystonia, polycythemia and cirrhosis
NM_018713.2(SLC30A10):c.460C>T (p.Gln154Ter)	SLC30A10	1	219927981	Hypermanganesemia with dystonia, polycythemia and cirrhosis
NM_006721.3(ADK):c.89G>A (p.Gly30Glu)	ADK	10	74200787	Hypermethioninemia due to adenosine kinase deficiency
NM_001161766.1(AHCY):c.252G>A (p.Trp84Ter)	AHCY	20	34292467	Hypermethioninemia with s-adenosylhomocysteine hydrolase deficiency
NM_014252.3(SLC25A15):c.538G>A (p.Glu180Lys)	SLC25A15	13	40807379	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_014252.3(SLC25A15):c.535C>T (p.Arg179Ter)	SLC25A15	13	40807376	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_014252.3(SLC25A15):c.79G>A (p.Gly27Arg)	SLC25A15	13	40799080	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_014252.3(SLC25A15):c.824G>A (p.Arg275Gln)	SLC25A15	13	40809585	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_014252.3(SLC25A15):c.815C>T (p.Thr272Ile)	SLC25A15	13	40809576	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_014252.3(SLC25A15):c.569G>A (p.Gly190Asp)	SLC25A15	13	40807410	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_014252.3(SLC25A15):c.658G>A (p.Gly220Arg)	SLC25A15	13	40808473	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_014252.3(SLC25A15):c.823C>T (p.Arg275Ter)	SLC25A15	13	40809584	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_014252.3(SLC25A15):c.847C>T (p.Leu283Phe)	SLC25A15	13	40809608	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
NM_130803.2(MEN1):c.1365+1G>A	MEN1	11	64805033	Hyperparathyroidism 1
NM_024529.4(CDC73):c.131+1G>A	CDC73	1	193122332	Hyperparathyroidism 1;Parathyroid adenoma, somatic;not provided
NM_024529.4(CDC73):c.3G>A (p.Met1Ile)	CDC73	1	193122203	Hyperparathyroidism 2
NM_024529.4(CDC73):c.25C>T (p.Arg9Ter)	CDC73	1	193122225	Hyperparathyroidism 2
NM_024529.4(CDC73):c.238-1G>A	CDC73	1	193130173	Hyperparathyroidism 2
NM_000317.2(PTS):c.46C>T (p.Arg16Cys)	PTS	11	112226489	Hyperphenylalaninemia, bh4-deficient, a, due to partial pts deficiency
NM_000281.3(PCBD1):c.236C>T (p.Thr79Ile)	PCBD1	10	70884029	Hyperphenylalaninemia, BH4-deficient, D
NM_000281.3(PCBD1):c.292C>T (p.Gln98Ter)	PCBD1	10	70883973	Hyperphenylalaninemia, BH4-deficient, D
NM_000277.1(PAH):c.997C>T (p.Leu333Phe)	PAH	12	102844404	Hyperphenylalaninemia, non-pku;not provided
NM_000277.1(PAH):c.140C>T (p.Ala47Val)	PAH	12	102912819	Hyperphenylalaninemia, non-pku;not provided
NM_000277.1(PAH):c.1243G>A (p.Asp415Asn)	PAH	12	102840472	Hyperphenylalaninemia, non-pku;Phenylketonuria;not provided
NM_000277.1(PAH):c.1139C>T (p.Thr380Met)	PAH	12	102843706	Hyperphenylalaninemia, non-pku;Phenylketonuria;not provided
NM_002546.3(TNFRSF11B):c.260G>A (p.Cys87Tyr)	TNFRSF11B	8	118933071	Hyperphosphatasemia with bone disease
NM_017837.3(PIGV):c.1022C>T (p.Ala341Val)	PIGV	1	26795056	Hyperphosphatasia with mental retardation syndrome 1
NM_017837.3(PIGV):c.467G>A (p.Cys156Tyr)	PIGV	1	26794501	Hyperphosphatasia with mental retardation syndrome 1;not provided

NM_032634.3(PIGO):c.2869C>T (p.Leu957Phe)	PIGO	9	35090266	Hyperphosphatasia with mental retardation syndrome 2
NM_032634.3(PIGO):c.3069+5G>A	PIGO	9	35090061	Hyperphosphatasia with mental retardation syndrome 2
NM_001256240.1(PGAP2):c.46C>T (p.Arg16Trp)	PGAP2	11	3811305	Hyperphosphatasia with mental retardation syndrome 3
NM_001145438.2(PGAP2):c.479C>T (p.Thr160Ile)	PGAP2	11	3824025	Hyperphosphatasia with mental retardation syndrome 3
NM_033419.4(PGAP3):c.275G>A (p.Gly92Asp)	PGAP3	17	39685926	Hyperphosphatasia with mental retardation syndrome 4
NM_033419.4(PGAP3):c.*559C>T	PGAP3	17	39672244	Hyperphosphatasia with mental retardation syndrome 4
NM_033419.4(PGAP3):c.558-10G>A	PGAP3	17	39673660	Hyperphosphatasia with mental retardation syndrome 4
NM_033419.4(PGAP3):c.320C>T (p.Ser107Leu)	PGAP3	17	39684709	Hyperphosphatasia with mental retardation syndrome 4
NM_001042616.2(PIGY):c.-540G>A	PIGY	4	88523797	Hyperphosphatasia with mental retardation syndrome 6
NM_000207.2(INS):c.266G>A (p.Arg89His)	INS	11	2159919	Hyperproinsulinemia
NM_000537.3(REN):c.1159C>T (p.Arg387Ter)	REN	1	204155078	Hyperproreninemia, familial
NM_004137.3(KCNMB1):c.193G>A (p.Glu65Lys)	KCNMB1	5	170383792	Hypertension, diastolic, resistance to
NM_000901.4(NR3C2):c.2429C>T (p.Ser810Leu)	NR3C2	4	148152550	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy
NM_000296.3(PKD1):c.8311G>A (p.Glu2771Lys)	PKD1	16	2103746	Hypertension;Multiple renal cysts
NM_000296.3(PKD1):c.3520C>T (p.Gln1174Ter)	PKD1	16	2111647	Hypertension;Polycystic kidney dysplasia
NM_000369.2(TSHR):c.2015G>A (p.Cys672Tyr)	TSHR	14	81144073	Hyperthyroidism, nonautoimmune
NM_000369.2(TSHR):c.1514G>A (p.Ser505Asn)	TSHR	14	81143572	Hyperthyroidism, nonautoimmune
NM_000369.2(TSHR):c.842G>A (p.Ser281Asn)	TSHR	14	81139828	Hyperthyroidism, nonautoimmune
NM_000369.2(TSHR):c.1915C>T (p.Pro639Ser)	TSHR	14	81143973	Hyperthyroidism, nonautoimmune
NM_000369.2(TSHR):c.1291G>A (p.Gly431Ser)	TSHR	14	81143349	Hyperthyroidism, nonautoimmune
NM_005691.3(ABCC9):c.3460C>T (p.Arg1154Trp)	ABCC9	12	21842327	Hypertrichotic osteochondrodysplasia
NM_005691.3(ABCC9):c.3128G>A (p.Cys1043Tyr)	ABCC9	12	21844884	Hypertrichotic osteochondrodysplasia
NM_005691.3(ABCC9):c.1433C>T (p.Ala478Val)	ABCC9	12	21908099	Hypertrichotic osteochondrodysplasia
NM_020297.3(ABCC9):c.3347G>A (p.Arg1116His)	ABCC9	12	21842440	Hypertrichotic osteochondrodysplasia
NM_005691.3(ABCC9):c.3346C>T (p.Arg1116Cys)	ABCC9	12	21842441	Hypertrichotic osteochondrodysplasia
NM_005691.3(ABCC9):c.178C>T (p.His60Tyr)	ABCC9	12	21933888	Hypertrichotic osteochondrodysplasia
NM_020297.3(ABCC9):c.3461G>A (p.Arg1154Gln)	ABCC9	12	21842326	Hypertrichotic osteochondrodysplasia;not provided
NM_000256.3(MYBPC3):c.906-36G>A	MYBPC3	11	47347065	Hypertrophic cardiomyopathy
NM_000363.4(TNNI3):c.407G>A (p.Arg136Gln)	TNNI3	19	55154172	Hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.2149-1G>A	MYBPC3	11	47338680	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.2454G>A (p.Trp818Ter)	MYBPC3	11	47337539	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.2670G>A (p.Trp890Ter)	MYBPC3	11	47335944	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3811C>T (p.Arg1271Ter)	MYBPC3	11	47332075	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.927-9G>A	MYBPC3	11	47346379	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.1987C>T (p.Arg663Cys)	MYH7	14	23426834	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000363.4(TNNI3):c.422G>A (p.Arg141Gln)	TNNI3	19	55154157	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000363.4(TNNI3):c.434G>A (p.Arg145Gln)	TNNI3	19	55154145	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000363.4(TNNI3):c.470C>T (p.Ala157Val)	TNNI3	19	55154109	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000363.4(TNNI3):c.557G>A (p.Arg186Gln)	TNNI3	19	55151910	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3190+5G>A	MYBPC3	11	47333552	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.1210C>T (p.Gln404Ter)	MYBPC3	11	47343505	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.966G>A (p.Trp322Ter)	MYBPC3	11	47346331	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000363.4(TNNI3):c.611G>A (p.Arg204His)	TNNI3	19	55151856	Hypertrophic cardiomyopathy;Primary familial hypertrophic cardiomyopathy;not provided
NM_000137.2(FAH):c.1062+5G>A	FAH	15	80180230	Hypertyrosinemia;Tyrosinemia type I;not provided
NM_001177800.1(ADIPOQ):c.334C>T (p.Arg112Cys)	ADIPOQ	3	186854303	Hypoadiponectinemia

NM_000384.2(APOB):c.1672C>T (p.Arg558Ter)	APOB	2	21028484	Hypobetalipoproteinemia, familial, 1
NM_000384.2(APOB):c.6835C>T (p.Gln2279Ter)	APOB	2	21010033	Hypobetalipoproteinemia, normotriglyceridemic
NM_000388.3(CASR):c.346G>A (p.Ala116Thr)	CASR	3	122257241	Hypocalcemia, autosomal dominant 1
NM_000388.3(CASR):c.452C>T (p.Thr151Met)	CASR	3	122257347	Hypocalcemia, autosomal dominant 1
NM_000388.3(CASR):c.571G>A (p.Glu191Lys)	CASR	3	122261606	Hypocalcemia, autosomal dominant 1
NM_000388.3(CASR):c.2459C>T (p.Ser820Phe)	CASR	3	122284413	Hypocalcemia, autosomal dominant 1
NM_000388.3(CASR):c.662C>T (p.Pro221Leu)	CASR	3	122261697	Hypocalcemia, autosomal dominant 1
NM_000388.3(CASR):c.1810G>A (p.Glu604Lys)	CASR	3	122283764	Hypocalcemia, autosomal dominant 1;not provided
NM_002067.4(GNA11):c.542G>A (p.Arg181Gln)	GNA11	19	3115009	Hypocalcemia, autosomal dominant 2
NM_002067.4(GNA11):c.178C>T (p.Arg60Cys)	GNA11	19	3110190	Hypocalcemia, autosomal dominant 2
NM_000388.3(CASR):c.2383C>T (p.Arg795Trp)	CASR	3	122284337	Hypocalciuric hypercalcemia, familial, type 1
NM_000388.3(CASR):c.680G>A (p.Arg227Gln)	CASR	3	122261715	Hypocalciuric hypercalcemia, familial, type 1
NM_000388.3(CASR):c.413C>T (p.Thr138Met)	CASR	3	122257308	Hypocalciuric hypercalcemia, familial, type 1
NM_000388.3(CASR):c.428G>A (p.Gly143Glu)	CASR	3	122257323	Hypocalciuric hypercalcemia, familial, type 1
NM_000388.3(CASR):c.196C>T (p.Arg66Cys)	CASR	3	122257091	Hypocalciuric hypercalcemia, familial, type 1
NM_000388.3(CASR):c.1657G>A (p.Gly553Arg)	CASR	3	122282161	Hypocalciuric hypercalcemia, familial, type 1
NM_000388.3(CASR):c.889G>A (p.Glu297Lys)	CASR	3	122261924	Hypocalciuric hypercalcemia, familial, type 1;Neonatal severe hyperparathyroidism
NM_000388.3(CASR):c.1942C>T (p.Arg648Ter)	CASR	3	122283896	Hypocalciuric hypercalcemia, familial, type 1;Neonatal severe hyperparathyroidism
NM_000388.3(CASR):c.554G>A (p.Arg185Gln)	CASR	3	122261589	Hypocalciuric hypercalcemia, familial, type 1;Neonatal severe hyperparathyroidism;not provided
NM_000388.3(CASR):c.73C>T (p.Arg25Ter)	CASR	3	122254262	Hypocalciuric hypercalcemia, familial, type 1;not provided
NM_004069.4(AP2S1):c.43C>T (p.Arg15Cys)	AP2S1	19	46846103	Hypocalciuric hypercalcemia, familial, type III
NM_004069.4(AP2S1):c.44G>A (p.Arg15His)	AP2S1	19	46846102	Hypocalciuric hypercalcemia, familial, type III
NM_001844.4(COL2A1):c.3220G>A (p.Gly1074Ser)	COL2A1	12	47977373	Hypochondrogenesis
NM_001844.4(COL2A1):c.2320G>A (p.Gly774Ser)	COL2A1	12	47982142	Hypochondrogenesis
NM_001844.4(COL2A1):c.3158G>A (p.Gly1053Glu)	COL2A1	12	47977607	Hypochondrogenesis
NM_000142.4(FGFR3):c.251C>T (p.Ser84Leu)	FGFR3	4	1799395	Hypochondroplasia
NM_000142.4(FGFR3):c.791C>T (p.Thr264Met)	FGFR3	4	1801886	Hypochondroplasia
NM_000142.4(FGFR3):c.597C>T (p.His199=)	FGFR3	4	1801518	Hypochondroplasia
NM_001174125.1(SLC11A2):c.1333C>T (p.Arg445Cys)	SLC11A2	12	50992291	Hypochromic microcytic anemia with iron overload
NM_021957.3(GYS2):c.736C>T (p.Arg246Ter)	GYS2	12	21568952	Hypoglycemia with deficiency of glycogen synthetase in the liver
NM_001059.2(TACR3):c.824G>A (p.Trp275Ter)	TACR3	4	103656258	Hypogonadotropic hypogonadism 11 with or without anosmia;Isolated GnRH Deficiency
NM_001059.2(TACR3):c.278G>A (p.Gly93Asp)	TACR3	4	103719398	Hypogonadotropic hypogonadism 11 with or without anosmia;not provided
NM_001059.2(TACR3):c.1057C>T (p.Pro353Ser)	TACR3	4	103591515	Hypogonadotropic hypogonadism 11 with or without anosmia;not provided
NM_018117.11(WDR11):c.1303G>A (p.Ala435Thr)	WDR11	10	120871178	Hypogonadotropic hypogonadism 14 with or without anosmia;not provided
NM_030964.3(SPRY4):c.910G>A (p.Val304Ile)	SPRY4	5	142314268	Hypogonadotropic hypogonadism 17 with or without anosmia
NM_001127496.1(SPRY4):c.46G>A (p.Val16Ile)	SPRY4	5	142315063	Hypogonadotropic hypogonadism 17 with or without anosmia
NM_017563.4(IL17RD):c.1403C>T (p.Ser468Leu)	IL17RD	3	57098300	Hypogonadotropic hypogonadism 18 with anosmia
NM_001946.3(DUSP6):c.545C>T (p.Ser182Phe)	DUSP6	12	89350881	Hypogonadotropic hypogonadism 19 with or without anosmia
NM_001946.3(DUSP6):c.1037C>T (p.Thr346Met)	DUSP6	12	89349363	Hypogonadotropic hypogonadism 19 with or without anosmia
NM_023110.2(FGFR1):c.2164C>T (p.Pro722Ser)	FGFR1	8	38414174	Hypogonadotropic hypogonadism 2 with anosmia
NM_003867.3(FGF17):c.530G>A (p.Arg177His)	FGF17	8	22048128	Hypogonadotropic hypogonadism 20 without anosmia
NM_001024613.3(FEZF1):c.832C>T (p.His278Tyr)	FEZF1	7	122303281	Hypogonadotropic hypogonadism 22 with anosmia
NM_144773.3(PROKR2):c.743G>A (p.Arg248Gln)	PROKR2	20	5302452	Hypogonadotropic hypogonadism 3 without anosmia
NM_000406.2(GNRHR):c.785G>A (p.Arg262Gln)	GNRHR	4	67740682	Hypogonadotropic hypogonadism 7 with or without anosmia
NM_000406.2(GNRHR):c.268G>A (p.Glu90Lys)	GNRHR	4	67754068	Hypogonadotropic hypogonadism 7 with or without anosmia

NM_000406.2(GNRHR):c.523_742del	GNRHR	4	67744788	Hypogonadotropic hypogonadism 7 with or without anosmia	
NM_000406.2(GNRHR):c.511G>A (p.Ala171Thr)	GNRHR	4	67753825	Hypogonadotropic hypogonadism 7 with or without anosmia	
NM_000406.2(GNRHR):c.959C>T (p.Pro320Leu)	GNRHR	4	67740508	Hypogonadotropic hypogonadism 7 with or without anosmia	
NM_000406.2(GNRHR):c.842C>T (p.Thr281Ile)	GNRHR	4	67740625	Hypogonadotropic hypogonadism 7 with or without anosmia	
NM_000406.2(GNRHR):c.806C>T (p.Thr269Met)	GNRHR	4	67740661	Hypogonadotropic hypogonadism 7 with or without anosmia	
NM_000406.2(GNRHR):c.416G>A (p.Arg139His)	GNRHR	4	67753920	Hypogonadotropic hypogonadism 7 with or without anosmia;Isolated GnRH Deficiency	
NM_032551.4(KISS1R):c.991C>T (p.Arg331Ter)	KISS1R	19	920542	Hypogonadotropic hypogonadism 8 without anosmia	
NM_001399.4(EDA):c.502+1G>A	EDA	X	69957133	Hypohidrotic ectodermal dysplasia	
NM_001399.4(EDA):c.766C>T (p.Gln256Ter)	EDA	X	70030493	Hypohidrotic ectodermal dysplasia	
NM_001399.4(EDA):c.991C>T (p.Gln331Ter)	EDA	X	70035424	Hypohidrotic ectodermal dysplasia	
NM_001399.4(EDA):c.396+1G>A	EDA	X	69616705	Hypohidrotic ectodermal dysplasia	
NM_001399.4(EDA):c.676C>T (p.Gln226Ter)	EDA	X	70028006	Hypohidrotic ectodermal dysplasia	
NM_001399.4(EDA):c.822G>A (p.Trp274Ter)	EDA	X	70033426	Hypohidrotic ectodermal dysplasia	
NM_003639.4(IKBKG):c.1207C>T (p.Gln403Ter)	IKBKG	X	154364408	Hypohidrotic ectodermal dysplasia with immune deficiency	
NM_001399.4(EDA):c.457C>T (p.Arg153Cys)	EDA	X	69957087	Hypohidrotic ectodermal dysplasia;not provided	
NM_001399.4(EDA):c.730C>T (p.Arg244Ter)	EDA	X	70029527	Hypohidrotic ectodermal dysplasia;not provided	
NM_001399.4(EDA):c.871G>A (p.Gly291Arg)	EDA	X	70033475	Hypohidrotic ectodermal dysplasia;not provided	
NM_001399.4(EDA):c.67C>T (p.Gln23Ter)	EDA	X	69616375	Hypohidrotic X-linked ectodermal dysplasia	
NM_001399.4(EDA):c.187G>A (p.Glu63Lys)	EDA	X	69616495	Hypohidrotic X-linked ectodermal dysplasia	
NM_001399.4(EDA):c.826C>T (p.Arg276Cys)	EDA	X	70033430	Hypohidrotic X-linked ectodermal dysplasia	
NM_001399.4(EDA):c.463C>T (p.Arg155Cys)	EDA	X	69957093	Hypohidrotic X-linked ectodermal dysplasia;Hypohidrotic ectodermal dysplasia;not provided	
NM_001399.4(EDA):c.467G>A (p.Arg156His)	EDA	X	69957097	Hypohidrotic X-linked ectodermal dysplasia;Hypohidrotic ectodermal dysplasia;not provided	
NM_001399.4(EDA):c.466C>T (p.Arg156Cys)	EDA	X	69957096	Hypohidrotic X-linked ectodermal dysplasia;not provided	
NM_001399.4(EDA):c.1045G>A (p.Ala349Thr)	EDA	X	70035478	Hypohidrotic X-linked ectodermal dysplasia;not provided	
NM_001626.5(AKT2):c.49G>A (p.Glu17Lys)	AKT2	19	40257052	Hypoinsulinemic hypoglycemia and hemihypertrophy	
NM_000334.4(SCN4A):c.3395G>A (p.Arg1132Gln)	SCN4A	17	63947091	Hypokalemic periodic paralysis 1;Hypokalemic periodic paralysis, type 2	
NM_000069.2(CACNA1S):c.3716G>A (p.Arg1239His)	CACNA1S	1	201053538	Hypokalemic periodic paralysis 1;not provided	
NM_000069.2(CACNA1S):c.1583G>A (p.Arg528His)	CACNA1S	1	201077915	Hypokalemic periodic paralysis 1;not provided	
NM_000334.4(SCN4A):c.3472C>T (p.Pro1158Ser)	SCN4A	17	63945608	Hypokalemic periodic paralysis, type 2	
NM_000334.4(SCN4A):c.664C>T (p.Arg222Trp)	SCN4A	17	63971201	Hypokalemic periodic paralysis, type 2	
NM_000334.4(SCN4A):c.3404G>A (p.Arg1135His)	SCN4A	17	63947082	Hypokalemic periodic paralysis, type 2;not provided	
NM_017662.4(TRPM6):c.1420C>T (p.Arg474Ter)	TRPM6	9	74812322	Hypomagnesemia 1, intestinal	
NM_017662.4(TRPM6):c.166C>T (p.Arg56Ter)	TRPM6	9	74842330	Hypomagnesemia 1, intestinal	
NM_017662.4(TRPM6):c.422C>T (p.Ser141Leu)	TRPM6	9	74840146	Hypomagnesemia 1, intestinal	
NM_017662.4(TRPM6):c.2667+1G>A	TRPM6	9	74788613	Hypomagnesemia 1, intestinal	
NM_021603.3(FXYD2):c.115G>A (p.Gly39Arg)	FXYD2	11	117822424	Hypomagnesemia 2, renal	
NM_001963.5(EGF):c.3209C>T (p.Pro1070Leu)	EGF	4	110004540	Hypomagnesemia 4, renal	
NM_148960.2(CLDN19):c.59G>A (p.Gly20Asp)	CLDN19	1	42740005	Hypomagnesemia 5, renal, with ocular involvement	
NM_017649.4(CNNM2):c.1703C>T (p.Thr568Ile)	CNNM2	10	103049788	Hypomagnesemia 6, renal	
NM_017649.4(CNNM2):c.364G>A (p.Glu122Lys)	CNNM2	10	102918844	Hypomagnesemia, seizures, and mental retardation	
NM_017649.4(CNNM2):c.1069G>A (p.Glu357Lys)	CNNM2	10	102919549	Hypomagnesemia, seizures, and mental retardation	
NM_007055.3(POLR3A):c.2015G>A (p.Gly672Glu)	POLR3A	10	78007761	Hypomyelinating leukodystrophy 7	
NM_007055.3(POLR3A):c.418C>T (p.Arg140Ter)	POLR3A	10	78025043	Hypomyelinating leukodystrophy 7	
NM_007055.3(POLR3A):c.3013C>T (p.Arg1005Cys)	POLR3A	10	77985961	Hypomyelinating leukodystrophy 7	
NM_007055.3(POLR3A):c.1114G>A (p.Asp372Asn)	POLR3A	10	78021617	Hypomyelinating leukodystrophy 7	

NM_007055.3(POLR3A):c.2171G>A (p.Cys724Tyr)	POLR3A	10	78004792	Hypomyelinating leukodystrophy 7	
NM_007055.3(POLR3A):c.3014G>A (p.Arg1005His)	POLR3A	10	77985960	Hypomyelinating leukodystrophy 7	
NM_007055.3(POLR3A):c.3991G>A (p.Ala1331Thr)	POLR3A	10	77980174	Hypomyelinating leukodystrophy 7	
NM_007055.3(POLR3A):c.4006C>T (p.Gln1336Ter)	POLR3A	10	77980159	Hypomyelinating leukodystrophy 7	
NM_007055.3(POLR3A):c.1909+18G>A	POLR3A	10	78009519	Hypomyelinating leukodystrophy 7;not provided	
NM_007055.3(POLR3A):c.2617-1G>A	POLR3A	10	77993368	Hypomyelinating leukodystrophy 7;not provided	
NM_018082.5(POLR3B):c.2303G>A (p.Arg768His)	POLR3B	12	106457147	Hypomyelinating leukodystrophy 8, with or without oligodontia and/or hypogonadotropic hypogonadism	
NM_018082.5(POLR3B):c.1648C>T (p.Arg550Ter)	POLR3B	12	106433739	Hypomyelinating leukodystrophy 8, with or without oligodontia and/or hypogonadotropic hypogonadism	
NM_032581.3(FAM126A):c.51+1G>A	FAM126A	7	22991060	Hypomyelination and Congenital Cataract	
NM_032581.3(FAM126A):c.415-1G>A	FAM126A	7	22976809	Hypomyelination and Congenital Cataract	
NM_001349.3(DARS):c.821C>T (p.Ala274Val)	DARS	2	135920591	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	
NM_001349.3(DARS):c.1379G>A (p.Arg460His)	DARS	2	135911174	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	
NM_001349.3(DARS):c.1459C>T (p.Arg487Cys)	DARS	2	135907363	Hypomyelination with brainstem and spinal cord involvement and leg spasticity;not provided	
NM_003705.4(SLC25A12):c.1058G>A (p.Arg353Gln)	SLC25A12	2	171813452	Hypomyelination, global cerebral	
NM_004752.3(GCM2):c.187G>A (p.Gly63Ser)	GCM2	6	10877296	Hypoparathyroidism familial isolated	
NM_000478.5(ALPL):c.648+1G>A	ALPL	1	21564217	Hypophosphatasia, perinatal lethal	
NM_000478.5(ALPL):c.1001G>A (p.Gly334Asp)	ALPL	1	21575736	Hypophosphatasia;Infantile hypophosphatasia;not provided	
NM_153638.3(PANK2):c.1441C>T (p.Arg481Ter)	PANK2	20	3916955	Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration	
NM_153638.3(PANK2):c.1561G>A (p.Gly521Arg)	PANK2	20	3918695	Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration;Inborn genetic diseases;Neurodegeneration with brain iron accumulation 1, atypical;Pigmentary pallidal degeneration;not provided	
NM_153638.3(PANK2):c.1583C>T (p.Thr528Met)	PANK2	20	3918717	Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration;Neurodegeneration with brain iron accumulation 1, atypical;Pigmentary pallidal degeneration	
NM_000044.4(AR):c.1645C>T (p.Pro549Ser)	AR	X	67643284	Hypospadias 1, X-linked	
NM_005491.4(MAMLD1):c.808C>T (p.Gln270Ter)	MAMLD1	X	150470381	Hypospadias 2, X-linked	
NM_005491.4(MAMLD1):c.2176C>T (p.Arg726Ter)	MAMLD1	X	150503409	Hypospadias 2, X-linked	
NM_000523.3(HOXD13):c.820C>T (p.Arg274Ter)	HOXD13	2	176094518	Hypospadias;Male infertility;Mesoaxial hand polydactyly;Oligospermia;Polydactyly;Syndactyly;not provided	
NM_001170961.1(IGSF1):c.2931G>A (p.Trp977Ter)	IGSF1	X	131275746	Hypothyroidism, central, and testicular enlargement	
NM_001170961.1(IGSF1):c.2588C>T (p.Ser863Phe)	IGSF1	X	131276974	Hypothyroidism, central, and testicular enlargement	
NM_000369.2(TSHR):c.326G>A (p.Arg109Gln)	TSHR	14	81087962	Hypothyroidism, congenital, nongoitrous, 1	
NM_000369.2(TSHR):c.970C>T (p.Gln324Ter)	TSHR	14	81143028	Hypothyroidism, congenital, nongoitrous, 1	
NM_000369.2(TSHR):c.1228G>A (p.Asp410Asn)	TSHR	14	81143286	Hypothyroidism, congenital, nongoitrous, 1	
NM_000369.2(TSHR):c.1430C>T (p.Thr477Ile)	TSHR	14	81143488	Hypothyroidism, congenital, nongoitrous, 1	
NM_000369.2(TSHR):c.928C>T (p.Arg310Cys)	TSHR	14	81142986	Hypothyroidism, congenital, nongoitrous, 1;not specified	
NM_199334.3(THRA):c.1207G>A (p.Glu403Lys)	THRA	17	40089430	Hypothyroidism, congenital, nongoitrous, 6	
NM_004473.3(FOXE1):c.304C>T (p.Arg102Cys)	FOXE1	9	97854218	Hypothyroidism, thyroidal, with spiky hair and cleft palate	
NM_001005463.2(EBF3):c.625C>T (p.Arg209Trp)	EBF3	10	129877779	HYPOTONIA, ATAXIA, AND DELAYED DEVELOPMENT SYNDROME	
NM_001005463.2(EBF3):c.616C>T (p.Arg206Ter)	EBF3	10	129877788	HYPOTONIA, ATAXIA, AND DELAYED DEVELOPMENT SYNDROME	
NM_052867.3(NALCN):c.1924C>T (p.Gln642Ter)	NALCN	13	101144812	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	
NM_032504.1(UNC80):c.5098C>T (p.Pro1700Ser)	UNC80	2	209918616	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	
NM_032504.1(UNC80):c.151C>T (p.Arg51Ter)	UNC80	2	209775898	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	

NM_001163436.2(TBCK):c.1532G>A (p.Arg511His)	TBCK	4	106233045	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3
NM_001163435.2(TBCK):c.376C>T (p.Arg126Ter)	TBCK	4	106262103	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3;not provided
NM_003094.3(SNRPE):c.133G>A (p.Gly45Ser)	SNRPE	1	203863714	Hypotrichosis 11
NM_000982.3(RPL21):c.95G>A (p.Arg32Gln)	RPL21	13	27254247	Hypotrichosis 12
NM_001264.4(CDSN):c.643C>T (p.Gln215Ter)	CDSN	6	31116972	Hypotrichosis 2
NM_001264.4(CDSN):c.598C>T (p.Gln200Ter)	CDSN	6	31117017	Hypotrichosis 2
NM_175053.3(KRT74):c.1444G>A (p.Asp482Asn)	KRT74	12	52567115	Hypotrichosis 3;not provided
NM_005144.4(HR):c.-315C>T	HR	8	22130702	Hypotrichosis 4
NM_177986.4(DSG4):c.865C>T (p.Arg289Ter)	DSG4	18	31392200	Hypotrichosis 6
NM_005767.5(LPAR6):c.463C>T (p.Gln155Ter)	LPAR6	13	48411961	Hypotrichosis 8
NM_005767.5(LPAR6):c.436G>A (p.Gly146Arg)	LPAR6	13	48411988	Hypotrichosis 8
NM_005767.5(LPAR6):c.565G>A (p.Glu189Lys)	LPAR6	13	48411859	Hypotrichosis 8
NM_005767.5(LPAR6):c.587C>T (p.Pro196Leu)	LPAR6	13	48411837	Hypotrichosis 8
NM_018419.2(SOX18):c.481C>T (p.Gln161Ter)	SOX18	20	64048840	Hypotrichosis-lymphedema-telangiectasia syndrome
NM_001491.2(GCNT2):c.1043G>A (p.Gly348Glu)	GCNT2	6	10626447	I blood group system
NM_001491.2(GCNT2):c.1148G>A (p.Arg383His)	GCNT2	6	10626552	I blood group system
NM_145649.4(GCNT2):c.505G>A (p.Ala169Thr)	GCNT2	6	10529416	I blood group system
NM_145649.4(GCNT2):c.683G>A (p.Arg228Gln)	GCNT2	6	10529594	I blood group system
NM_024312.4(GNPTAB):c.310C>T (p.Gln104Ter)	GNPTAB	12	101789951	I cell disease
NM_024312.4(GNPTAB):c.2681G>A (p.Trp894Ter)	GNPTAB	12	101764236	I cell disease
NM_024312.4(GNPTAB):c.3335+1G>A	GNPTAB	12	101757571	I cell disease
NM_024312.4(GNPTAB):c.2533C>T (p.Gln845Ter)	GNPTAB	12	101764384	I cell disease
NM_024312.4(GNPTAB):c.3249+1G>A	GNPTAB	12	101760029	I cell disease
NM_024312.4(GNPTAB):c.3434+1G>A	GNPTAB	12	101757211	I cell disease
NM_024312.4(GNPTAB):c.1090C>T (p.Arg364Ter)	GNPTAB	12	101770429	I cell disease
NM_024312.4(GNPTAB):c.136C>T (p.Arg46Ter)	GNPTAB	12	101796744	I cell disease
NM_024312.4(GNPTAB):c.1519C>T (p.Gln507Ter)	GNPTAB	12	101766184	I cell disease
NM_024312.4(GNPTAB):c.1759C>T (p.Arg587Ter)	GNPTAB	12	101765158	I cell disease
NM_024312.4(GNPTAB):c.3061C>T (p.Gln1021Ter)	GNPTAB	12	101761201	I cell disease
NM_024312.4(GNPTAB):c.3091C>T (p.Arg1031Ter)	GNPTAB	12	101761171	I cell disease
NM_024312.4(GNPTAB):c.637-1G>A	GNPTAB	12	101780287	I cell disease
NM_024312.4(GNPTAB):c.940C>T (p.Gln314Ter)	GNPTAB	12	101770579	I cell disease
NM_024312.4(GNPTAB):c.1298G>A (p.Trp433Ter)	GNPTAB	12	101768147	I cell disease;not provided
NM_024312.4(GNPTAB):c.3565C>T (p.Arg1189Ter)	GNPTAB	12	101753409	I cell disease;Pseudo-Hurler polydystrophy
NM_024312.4(GNPTAB):c.3613C>T (p.Arg1205Ter)	GNPTAB	12	101749181	I cell disease;Pseudo-Hurler polydystrophy
NM_024312.4(GNPTAB):c.1000C>T (p.Arg334Ter)	GNPTAB	12	101770519	I cell disease;Pseudo-Hurler polydystrophy;not provided
NM_000423.2(KRT2):c.1459G>A (p.Glu487Lys)	KRT2	12	52646750	Ichthyosis bullosa of Siemens;Ichthyosis exfoliativa;not provided
NM_000423.2(KRT2):c.1426G>A (p.Glu476Lys)	KRT2	12	52646783	Ichthyosis bullosa of Siemens;not provided
NM_005094.3(SLC27A4):c.274G>A (p.Ala92Thr)	SLC27A4	9	128345267	Ichthyosis prematurity syndrome
NM_005094.3(SLC27A4):c.1748G>A (p.Arg583His)	SLC27A4	9	128355770	Ichthyosis prematurity syndrome
NM_002016.1(FLG):c.2143C>T (p.Gln715Ter)	FLG	1	152312743	Ichthyosis vulgaris
NM_002016.1(FLG):c.1501C>T (p.Arg501Ter)	FLG	1	152313385	Ichthyosis vulgaris;not provided
NM_021978.3(ST14):c.2479G>A (p.Gly827Arg)	ST14	11	130209734	Ichthyosis, congenital, autosomal recessive 11
NM_021978.3(ST14):c.3G>A (p.Met1Ile)	ST14	11	130159982	Ichthyosis, congenital, autosomal recessive 11
NM_021978.3(ST14):c.2269+1G>A	ST14	11	130208685	Ichthyosis, congenital, autosomal recessive 11

NM_022726.3(ELOVL4):c.646C>T (p.Arg216Ter)	ELOVL4	6	79919443	ichthyosis, spastic quadriplegia, and mental retardation
NM_001257180.1(SLC20A2):c.1802C>T (p.Ser601Leu)	SLC20A2	8	42417960	Idiopathic basal ganglia calcification 1
NM_006749.4(SLC20A2):c.1723G>A (p.Glu575Lys)	SLC20A2	8	42428829	Idiopathic basal ganglia calcification 1
NM_006749.4(SLC20A2):c.1784C>T (p.Thr595Met)	SLC20A2	8	42428768	Idiopathic basal ganglia calcification 1
NM_002608.3(PDGFB):c.433C>T (p.Gln145Ter)	PDGFB	22	39231645	Idiopathic basal ganglia calcification 5
NM_002608.3(PDGFB):c.445C>T (p.Arg149Ter)	PDGFB	22	39231633	Idiopathic basal ganglia calcification 5
NM_002608.3(PDGFB):c.3G>A (p.Met1Ile)	PDGFB	22	39243961	Idiopathic basal ganglia calcification 5
NM_198253.2(TERT):c.1456C>T (p.Arg486Cys)	TERT	5	1293430	Idiopathic fibrosing alveolitis, chronic form
NM_198253.2(TERT):c.97C>T (p.Pro33Ser)	TERT	5	1294893	Idiopathic fibrosing alveolitis, chronic form
NM_016434.3(RTEL1):c.1482-1G>A	RTEL1	20	63687936	Idiopathic fibrosing alveolitis, chronic form
NM_198253.2(TERT):c.430G>A (p.Val144Met)	TERT	5	1294456	Idiopathic fibrosing alveolitis, chronic form;not provided
NM_198253.2(TERT):c.219+1G>A	TERT	5	1294770	Idiopathic fibrosing alveolitis, chronic form;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1
NM_198253.2(TERT):c.2594G>A (p.Arg865His)	TERT	5	1266524	Idiopathic fibrosing alveolitis, chronic form;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1;not provided
NR_001566.1(TERC):n.98G>A	TERC	3	169764963	Idiopathic fibrosing alveolitis, chronic form;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 2
NM_016434.3(RTEL1):c.2957G>A (p.Arg986Gln)	RTEL1	20	63693248	Idiopathic fibrosing alveolitis, chronic form;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3
NM_000782.4(CYP24A1):c.476G>A (p.Arg159Gln)	CYP24A1	20	54171644	Idiopathic hypercalcemia of infancy
NM_000782.4(CYP24A1):c.964G>A (p.Glu322Lys)	CYP24A1	20	54162743	Idiopathic hypercalcemia of infancy
NM_000782.4(CYP24A1):c.1186C>T (p.Arg396Trp)	CYP24A1	20	54158136	Idiopathic hypercalcemia of infancy;Muscle cramps
NM_177405.2(ADA2):c.424G>A (p.Gly142Ser)	ADA2	22	17182696	Idiopathic livedo reticularis with systemic involvement
NM_000203.4(IDUA):c.898G>A (p.Ala300Thr)	IDUA	4	1002087	IDUA pseudodeficiency
NM_015884.3(MBTPS2):c.261G>A (p.Met87Ile)	MBTPS2	X	21845207	IFAP syndrome with or without BRESHECK syndrome
NM_015884.3(MBTPS2):c.1286G>A (p.Arg429His)	MBTPS2	X	21880921	IFAP syndrome with or without BRESHECK syndrome;not provided
NM_005842.3(SPRY2):c.355C>T (p.Arg119Trp)	SPRY2	13	80337351	IgA nephropathy, susceptibility to, 3
NM_000566.3(FCGR1A):c.274C>T (p.Arg92Ter)	FCGR1A	1	149784224	IGG receptor I, phagocytic, familial deficiency of
NM_032790.3(ORAI1):c.271C>T (p.Arg91Trp)	ORAI1	12	121627018	Immune dysfunction with T-cell inactivation due to calcium entry defect 1
NM_003156.3(STIM1):c.970-1G>A	STIM1	11	4082183	Immune dysfunction with T-cell inactivation due to calcium entry defect 2
NM_003156.3(STIM1):c.1285C>T (p.Arg429Cys)	STIM1	11	4083309	Immune dysfunction with T-cell inactivation due to calcium entry defect 2
NM_003156.3(STIM1):c.1276C>T (p.Arg426Cys)	STIM1	11	4083300	Immune dysfunction with T-cell inactivation due to calcium entry defect 2
NM_032415.5(CARD11):c.2833C>T (p.Gln945Ter)	CARD11	7	2915243	Immunodeficiency 11
NM_005026.4(PIK3CD):c.1573G>A (p.Glu525Lys)	PIK3CD	1	9720793	Immunodeficiency 14
NM_005026.4(PIK3CD):c.3061G>A (p.Glu1021Lys)	PIK3CD	1	9726972	Immunodeficiency 14;not provided
NM_001556.2(IKBKB):c.814C>T (p.Arg272Ter)	IKBKB	8	42316223	Immunodeficiency 15
NM_000733.3(CD3E):c.176G>A (p.Trp59Ter)	CD3E	11	118312690	Immunodeficiency 18
NM_000732.4(CD3D):c.202C>T (p.Arg68Ter)	CD3D	11	118340447	Immunodeficiency 19
NM_000732.4(CD3D):c.274+5G>A	CD3D	11	118340370	Immunodeficiency 19
NM_006904.6(PRKDC):c.10721C>T (p.Ala3574Val)	PRKDC	8	47789188	Immunodeficiency 26 with or without neurologic abnormalities
NM_005535.2(IL12RB1):c.94C>T (p.Gln32Ter)	IL12RB1	19	18083462	Immunodeficiency 30
NM_005535.2(IL12RB1):c.1126C>T (p.Gln376Ter)	IL12RB1	19	18069609	Immunodeficiency 30
NM_005535.2(IL12RB1):c.637C>T (p.Arg213Trp)	IL12RB1	19	18075812	Immunodeficiency 30
NM_007315.3(STAT1):c.820C>T (p.Arg274Trp)	STAT1	2	190995185	Immunodeficiency 31C
NM_007315.3(STAT1):c.800C>T (p.Ala267Val)	STAT1	2	190995205	Immunodeficiency 31C

NM_007315.3(STAT1):c.821G>A (p.Arg274Gln)	STAT1	2	190995184	Immunodeficiency 31C	
NM_007315.3(STAT1):c.1154C>T (p.Thr385Met)	STAT1	2	190986921	Immunodeficiency 31C	
NM_007315.3(STAT1):c.1151G>A (p.Gly384Asp)	STAT1	2	190986924	Immunodeficiency 31C	
NM_003921.4(BCL10):c.57+1G>A	BCL10	1	85276295	Immunodeficiency 37	
NM_005101.3(ISG15):c.163C>T (p.Gln55Ter)	ISG15	1	1014143	Immunodeficiency 38 with basal ganglia calcification	
NM_001572.3(IRF7):c.1261C>T (p.Gln421Ter)	IRF7	11	613094	Immunodeficiency 39	
NM_004946.2(DOCK2):c.3970C>T (p.Gln1324Ter)	DOCK2	5	170047513	Immunodeficiency 40	
NM_004946.2(DOCK2):c.3310C>T (p.Arg1104Trp)	DOCK2	5	170019037	Immunodeficiency 40	
NM_005060.3(RORC):c.113C>T (p.Ser38Leu)	RORC	1	151817238	Immunodeficiency 42	
NM_001001523.1(RORC):c.922C>T (p.Gln308Ter)	RORC	1	151813569	Immunodeficiency 42	
NM_001001523.1(RORC):c.1258C>T (p.Gln420Ter)	RORC	1	151811399	Immunodeficiency 42	
NM_002444.2(MSN):c.511C>T (p.Arg171Trp)	MSN	X	65731150	IMMUNODEFICIENCY 50	
NM_002444.2(MSN):c.1657C>T (p.Arg553Ter)	MSN	X	65739816	IMMUNODEFICIENCY 50	
NM_001193333.2(CORO1A):c.400G>A (p.Val134Met)	CORO1A	16	30186894	Immunodeficiency 8	
NM_198053.2(CD247):c.208C>T (p.Gln70Ter)	CD247	1	167439355	Immunodeficiency due to defect in cd3-zeta	
NM_001183.5(ATP6AP1):c.1284G>A (p.Met428Ile)	ATP6AP1	X	154435762	Immunodeficiency due to ficolin 3 deficiency	
NM_001183.5(ATP6AP1):c.1036G>A (p.Glu346Lys)	ATP6AP1	X	154435338	Immunodeficiency due to ficolin 3 deficiency	
NM_000074.2(CD40LG):c.419G>A (p.Trp140Ter)	CD40LG	X	136659048	Immunodeficiency with hyper IgM type 1	
NM_000074.2(CD40LG):c.31C>T (p.Arg11Ter)	CD40LG	X	136648279	Immunodeficiency with hyper IgM type 1	
NM_000074.2(CD40LG):c.761C>T (p.Thr254Met)	CD40LG	X	136659390	Immunodeficiency with hyper IgM type 1	
NM_020661.3(AICDA):c.70C>T (p.Arg24Trp)	AICDA	12	8606951	Immunodeficiency with hyper IgM type 2	
NM_020661.3(AICDA):c.203G>A (p.Trp68Ter)	AICDA	12	8605439	Immunodeficiency with hyper IgM type 2	
NM_020661.3(AICDA):c.251G>A (p.Trp84Ter)	AICDA	12	8605391	Immunodeficiency with hyper IgM type 2;not provided	
NM_006060.5(IKZF1):c.485G>A (p.Arg162Gln)	IKZF1	7	50382603	Immunodeficiency, common variable, 13	
NM_006060.5(IKZF1):c.551G>A (p.Arg184Gln)	IKZF1	7	50382669	Immunodeficiency, common variable, 13	
NM_032121.5(MAGT1):c.409C>T (p.Arg137Ter)	MAGT1	X	77870885	Immunodeficiency, X-Linked, with magnesium defect, Epstein-Barr virus infection, and neoplasia	
NM_014797.2(ZBTB24):c.958C>T (p.Arg320Ter)	ZBTB24	6	109476925	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	
NM_014797.2(ZBTB24):c.1369C>T (p.Arg457Ter)	ZBTB24	6	109467654	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	
NM_031942.4(CDCA7):c.1057C>T (p.Arg353Cys)	CDCA7	2	173366304	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	
NM_031942.4(CDCA7):c.1148G>A (p.Arg383His)	CDCA7	2	173366395	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	
NM_031942.4(CDCA7):c.1058G>A (p.Arg353His)	CDCA7	2	173366305	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	
NM_001440.3(EXTL3):c.1382C>T (p.Pro461Leu)	EXTL3	8	28717441	IMMUNOSKELETAL DYSPLASIA WITH NEURODEVELOPMENTAL ABNORMALITIES	
NM_005850.4(SF3B4):c.164-1G>A	SF3B4	1	149926919	Inborn genetic diseases	
NM_006922.3(SCN3A):c.1318C>T (p.Gln440Ter)	SCN3A	2	165154514	Inborn genetic diseases	
NM_020919.3(ALS2):c.4897C>T (p.Gln1633Ter)	ALS2	2	201704160	Inborn genetic diseases	
NM_002047.3(GARS):c.373G>A (p.Glu125Lys)	GARS	7	30599995	Inborn genetic diseases	
NM_014112.4(TRPS1):c.2627C>T (p.Ser876Phe)	TRPS1	8	115587074	Inborn genetic diseases	
NM_007327.3(GRIN1):c.2443G>A (p.Gly815Arg)	GRIN1	9	137163668	Inborn genetic diseases	
NM_000834.3(GRIN2B):c.1916C>T (p.Ala639Val)	GRIN2B	12	13608697	Inborn genetic diseases	
NM_000217.2(KCNA1):c.1202C>T (p.Ala401Val)	KCNA1	12	4912580	Inborn genetic diseases	
NM_001845.5(COL4A1):c.2494G>A (p.Gly832Arg)	COL4A1	13	110178196	Inborn genetic diseases	
NM_001376.4(DYNC1H1):c.10033G>A (p.Glu3345Lys)	DYNC1H1	14	102032421	Inborn genetic diseases	
NM_001271.3(CHD2):c.5035C>T (p.Arg1679Ter)	CHD2	15	93020140	Inborn genetic diseases	
NM_024306.4(FA2H):c.565C>T (p.Arg189Ter)	FA2H	16	74726273	Inborn genetic diseases	
NM_013275.5(ANKRD11):c.7825C>T (p.Gln2609Ter)	ANKRD11	16	89268645	Inborn genetic diseases	

NM_001042492.2(NF1):c.4729C>T (p.Gln1577Ter)	NF1	17	31265233	Inborn genetic diseases
NM_002501.3(NFIX):c.361C>T (p.Arg121Cys)	NFIX	19	13025354	Inborn genetic diseases
NM_003588.3(CUL4B):c.1906+1G>A	CUL4B	X	120538659	Inborn genetic diseases
NM_005334.2(HCFC1):c.5860G>A (p.Gly1954Arg)	HCFC1	X	153950387	Inborn genetic diseases
NM_003159.2(CDKL5):c.514G>A (p.Val172Ile)	CDKL5	X	18584313	Inborn genetic diseases
NM_000292.2(PHKA2):c.884G>A (p.Arg295His)	PHKA2	X	18940029	Inborn genetic diseases
NM_006306.3(SMC1A):c.3196C>T (p.Arg1066Cys)	SMC1A	X	53382595	Inborn genetic diseases
NM_018684.3(ZC4H2):c.148C>T (p.Gln50Ter)	ZC4H2	X	64921894	Inborn genetic diseases
NM_002857.3(PEX19):c.775C>T (p.Gln259Ter)	PEX19	1	160279842	Inborn genetic diseases
NM_018122.4(DARS2):c.127+1G>A	DARS2	1	173825357	Inborn genetic diseases
NM_000158.3(GBE1):c.1064G>A (p.Arg355His)	GBE1	3	81593952	Inborn genetic diseases
NM_006772.2(SYNGAP1):c.3718C>T (p.Arg1240Ter)	SYNGAP1	6	33446710	Inborn genetic diseases
NM_003130.3(SRI):c.206-1G>A	SRI	7	88210926	Inborn genetic diseases
NM_017890.4(VPS13B):c.2410C>T (p.Gln804Ter)	VPS13B	8	99192952	Inborn genetic diseases
NM_017890.4(VPS13B):c.9547C>T (p.Gln3183Ter)	VPS13B	8	99832510	Inborn genetic diseases
NM_005094.3(SLC27A4):c.1774G>A (p.Gly592Arg)	SLC27A4	9	128355796	Inborn genetic diseases
NM_052867.3(NALCN):c.4197+1G>A	NALCN	13	101073583	Inborn genetic diseases
NM_052867.3(NALCN):c.2203C>T (p.Arg735Ter)	NALCN	13	101111216	Inborn genetic diseases
NM_015346.3(ZFYVE26):c.7195C>T (p.Gln2399Ter)	ZFYVE26	14	67752520	Inborn genetic diseases
NM_003384.2(VRK1):c.1068+1G>A	VRK1	14	96860736	Inborn genetic diseases
NM_006031.5(PCNT):c.2728C>T (p.Gln910Ter)	PCNT	21	46366702	Inborn genetic diseases
NM_000254.2(MTR):c.3518C>T (p.Pro1173Leu)	MTR	1	236895470	Inborn genetic diseases;Intellectual disability, profound;METHYLCOBALAMIN DEFICIENCY, cblG TYPE;Seizure disorder;not provided
NM_181714.3(LCA5):c.835C>T (p.Gln279Ter)	LCA5	6	79493636	Inborn genetic diseases;Leber congenital amaurosis;Leber congenital amaurosis 5
NM_006009.3(TUBA1A):c.5G>A (p.Arg2His)	TUBA1A	12	49186832	Inborn genetic diseases;Lissencephaly 3
NM_005027.3(PIK3R2):c.1117G>A (p.Gly373Arg)	PIK3R2	19	18162974	Inborn genetic diseases;Megalencephaly cutis marmorata telangiectatica congenita;Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1;not provided
NM_001242875.2(ELP2):c.1579C>T (p.Arg527Trp)	ELP2	18	36156574	Inborn genetic diseases;MENTAL RETARDATION, AUTOSOMAL RECESSIVE 58;not provided
NM_003384.2(VRK1):c.1072C>T (p.Arg358Ter)	VRK1	14	96876033	Inborn genetic diseases;Multiple congenital anomalies;Pontocerebellar hypoplasia type 1
NM_018026.3(PACS1):c.607C>T (p.Arg203Trp)	PACS1	11	66211206	Inborn genetic diseases;Multiple congenital anomalies;Schuurs-hoeijmakers syndrome;not provided
NM_000489.4(ATRX):c.736C>T (p.Arg246Cys)	ATRX	X	77684520	Inborn genetic diseases;not provided
NM_019109.4(ALG1):c.1079C>T (p.Ala360Val)	ALG1	16	5082565	Inborn genetic diseases;not provided
NM_015265.3(SATB2):c.847C>T (p.Arg283Ter)	SATB2	2	199349027	Inborn genetic diseases;not provided
NM_006624.5(ZMYND11):c.1798C>T (p.Arg600Trp)	ZMYND11	10	252459	Inborn genetic diseases;not provided
NM_001376.4(DYNC1H1):c.4700G>A (p.Arg1567Gln)	DYNC1H1	14	102002694	Inborn genetic diseases;not provided
NM_019066.4(MAGEL2):c.1912C>T (p.Gln638Ter)	MAGEL2	15	23645831	Inborn genetic diseases;not provided
NM_138736.2(GNAO1):c.626G>A (p.Arg209His)	GNAO1	16	56336763	Inborn genetic diseases;not provided
NM_020988.2(GNAO1):c.736G>A (p.Glu246Lys)	GNAO1	16	56351396	Inborn genetic diseases;not provided
NM_013275.5(ANKRD11):c.7569+1G>A	ANKRD11	16	89275092	Inborn genetic diseases;not provided
NM_004977.2(KCNC3):c.1268G>A (p.Arg423His)	KCNC3	19	50323685	Inborn genetic diseases;not provided
NM_003491.3(NAA10):c.247C>T (p.Arg83Cys)	NAA10	X	153932410	Inborn genetic diseases;N-terminal acetyltransferase deficiency;not provided
NM_004595.4(SMS):c.166G>A (p.Gly56Ser)	SMS	X	21967312	Inborn genetic diseases;Snyder Robinson syndrome;not provided
NM_004820.4(CYP7B1):c.334C>T (p.Arg112Ter)	CYP7B1	8	64616207	Inborn genetic diseases;Spastic paraplegia
NM_015915.4(ATL1):c.1243C>T (p.Arg415Trp)	ATL1	14	50628154	Inborn genetic diseases;Spastic paraplegia 3
NM_014780.4(CUL7):c.4717C>T (p.Arg1573Ter)	CUL7	6	43038323	Inborn genetic diseases;Three M syndrome 1

NM_001615.3(ACTG2):c.533G>A (p.Arg178His)	ACTG2	2	73913566	Inborn genetic diseases;Visceral myopathy
NM_001128227.2(GNE):c.2008C>T (p.Gln670Ter)	GNE	9	36218201	Inclusion body myopathy 2
NM_001128227.2(GNE):c.268C>T (p.Arg90Ter)	GNE	9	36246472	Inclusion body myopathy 2
NM_001128227.2(GNE):c.830G>A (p.Arg277Gln)	GNE	9	36236864	Inclusion body myopathy 2;Nonaka myopathy
NM_005476.5(GNE):c.2086G>A (p.Val696Met)	GNE	9	36217448	Inclusion body myopathy 2;Nonaka myopathy;Sialuria;not provided
NM_001128227.2(GNE):c.1985C>T (p.Ala662Val)	GNE	9	36218224	Inclusion body myopathy 2;Nonaka myopathy;Sialuria;not provided
NM_001128227.2(GNE):c.829C>T (p.Arg277Trp)	GNE	9	36236865	Inclusion body myopathy 2;Sialuria
NM_017534.5(MYH2):c.2116G>A (p.Glu706Lys)	MYH2	17	10535137	Inclusion body myopathy 3
NM_017534.5(MYH2):c.904+1G>A	MYH2	17	10542874	Inclusion body myopathy 3
NM_017534.5(MYH2):c.2347C>T (p.Arg783Ter)	MYH2	17	10533379	Inclusion body myopathy 3
NM_017534.5(MYH2):c.706G>A (p.Ala236Thr)	MYH2	17	10543746	Inclusion body myopathy 3
NM_017534.5(MYH2):c.533C>T (p.Thr178Ile)	MYH2	17	10544100	Inclusion body myopathy 3
NM_007126.3(VCP):c.463C>T (p.Arg155Cys)	VCP	9	35065364	Inclusion body myopathy with early-onset paget disease and frontotemporal dementia;not provided
NM_007126.3(VCP):c.476G>A (p.Arg159His)	VCP	9	35065351	Inclusion body myopathy with early-onset paget disease and frontotemporal dementia;not provided
NM_003639.4(IKBKG):c.184C>T (p.Arg62Ter)	IKBKG	X	154552186	Incontinentia pigmenti syndrome;not provided
NM_002977.3(SCN9A):c.2691G>A (p.Trp897Ter)	SCN9A	2	166277133	Indifference to pain, congenital, autosomal recessive
NM_002977.3(SCN9A):c.829C>T (p.Arg277Ter)	SCN9A	2	166303162	Indifference to pain, congenital, autosomal recessive
NM_002977.3(SCN9A):c.2690G>A (p.Trp897Ter)	SCN9A	2	166277134	Indifference to pain, congenital, autosomal recessive
NM_001098.2(ACO2):c.776G>A (p.Gly259Asp)	ACO2	22	41515858	Infantile cerebellar-retinal degeneration
NM_145239.2(PRRT2):c.950G>A (p.Ser317Asn)	PRRT2	16	29814403	Infantile convulsions and paroxysmal choreoathetosis, familial
NM_145239.2(PRRT2):c.562C>T (p.Gln188Ter)	PRRT2	16	29813616	Infantile convulsions and paroxysmal choreoathetosis, familial
NM_145239.2(PRRT2):c.718C>T (p.Arg240Ter)	PRRT2	16	29813772	Infantile convulsions and paroxysmal choreoathetosis, familial;not provided
NM_000088.3(COL1A1):c.3040C>T (p.Arg1014Cys)	COL1A1	17	50188908	Infantile cortical hyperostosis;not provided
NM_000404.3(GLB1):c.1369C>T (p.Arg457Ter)	GLB1	3	33016819	Infantile GM1 gangliosidosis
NM_000404.3(GLB1):c.203G>A (p.Arg68Gln)	GLB1	3	33072586	Infantile GM1 gangliosidosis
NM_000404.3(GLB1):c.367G>A (p.Gly123Arg)	GLB1	3	33068849	Infantile GM1 gangliosidosis;not provided
m.15498G>A	MT-CYB	MT	15498	Infantile histiocytoid cardiomyopathy
NM_019056.6(NDUFB11):c.262C>T (p.Arg88Ter)	NDUFB11	X	47142690	Infantile histiocytoid cardiomyopathy;Linear skin defects with multiple congenital anomalies 3;not provided
NM_000478.5(ALPL):c.535G>A (p.Ala179Thr)	ALPL	1	21564103	Infantile hypophosphatasia
NM_000478.5(ALPL):c.211C>T (p.Arg71Cys)	ALPL	1	21561126	Infantile hypophosphatasia
NM_000478.5(ALPL):c.98C>T (p.Ala33Val)	ALPL	1	21560662	Infantile hypophosphatasia
NM_000478.5(ALPL):c.892G>A (p.Glu298Lys)	ALPL	1	21573694	Infantile hypophosphatasia
NM_000478.5(ALPL):c.1366G>A (p.Gly456Arg)	ALPL	1	21577439	Infantile hypophosphatasia
NM_015909.3(NBAS):c.409C>T (p.Arg137Trp)	NBAS	2	15539327	Infantile liver failure syndrome 2
NM_002609.3(PDGFRB):c.1681C>T (p.Arg561Cys)	PDGFRB	5	150125571	Infantile myofibromatosis;Infantile myofibromatosis 1;not provided
NM_014425.4(INVS):c.1807C>T (p.Arg603Ter)	INVS	9	100284342	Infantile nephronophthisis
NM_014425.4(INVS):c.2719C>T (p.Arg907Ter)	INVS	9	100292976	Infantile nephronophthisis
NM_014425.4(INVS):c.2509C>T (p.Gln837Ter)	INVS	9	100292766	Infantile nephronophthisis
NM_014425.4(INVS):c.2695C>T (p.Arg899Ter)	INVS	9	100292952	Infantile nephronophthisis;Nephronophthisis
NM_003560.3(PLA2G6):c.1117G>A (p.Gly373Arg)	PLA2G6	22	38129523	Infantile neuroaxonal dystrophy;Iron accumulation in brain
NM_003560.3(PLA2G6):c.109C>T (p.Arg37Ter)	PLA2G6	22	38169318	Infantile neuroaxonal dystrophy;Iron accumulation in brain;not provided
NM_194277.2(FRMD7):c.601C>T (p.Gln201Ter)	FRMD7	X	132085625	Infantile nystagmus, X-linked
NM_194277.2(FRMD7):c.1003C>T (p.Arg335Ter)	FRMD7	X	132080053	Infantile nystagmus, X-linked
NM_194277.2(FRMD7):c.252G>A (p.Val84=)	FRMD7	X	132097298	Infantile nystagmus, X-linked

NM_194277.2(FRMD7):c.70G>A (p.Gly24Arg)	FRMD7	X	132100704	Infantile nystagmus, X-linked
NM_194277.2(FRMD7):c.812G>A (p.Cys271Tyr)	FRMD7	X	132082456	Infantile nystagmus, X-linked
NM_001044.4(SLC6A3):c.1184C>T (p.Pro395Leu)	SLC6A3	5	1411328	Infantile Parkinsonism-dystonia
NM_001044.4(SLC6A3):c.1269+1G>A	SLC6A3	5	1411242	Infantile Parkinsonism-dystonia
NM_001044.4(SLC6A3):c.1031+1G>A	SLC6A3	5	1416097	Infantile Parkinsonism-dystonia
NM_001044.4(SLC6A3):c.1561C>T (p.Arg521Trp)	SLC6A3	5	1406226	Infantile Parkinsonism-dystonia
NM_000548.4(TSC2):c.4662+1G>A	TSC2	16	2085323	Infantile spasms;Intellectual disability, severe;Tuberous sclerosis syndrome
NM_020919.3(ALS2):c.2992C>T (p.Arg998Ter)	ALS2	2	201726854	Infantile-onset ascending hereditary spastic paralysis
NM_020919.3(ALS2):c.470G>A (p.Cys157Tyr)	ALS2	2	201761524	Infantile-onset ascending hereditary spastic paralysis
NM_020919.3(ALS2):c.2143C>T (p.Gln715Ter)	ALS2	2	201744285	Infantile-onset ascending hereditary spastic paralysis
NM_020919.3(ALS2):c.2761C>T (p.Arg921Ter)	ALS2	2	201728592	Infantile-onset ascending hereditary spastic paralysis;not provided
NM_001015879.1(AURKC):c.629G>A (p.Cys210Tyr)	AURKC	19	57234985	Infertility associated with multi-tailed spermatozoa and excessive DNA
NM_144701.2(IL23R):c.1142G>A (p.Arg381Gln)	IL23R	1	67240275	Inflammatory bowel disease 17, protection against;Psoriasis, protection against
NM_001145805.1(IRGM):c.313C>T (p.Leu105=)	IRGM	5	150848436	Inflammatory bowel disease 19
NM_000628.4(IL10RB):c.477G>A (p.Trp159Ter)	IL10RB	21	33279897	Inflammatory bowel disease 25, autosomal recessive
NM_001558.3(IL10RA):c.421G>A (p.Gly141Arg)	IL10RA	11	117993294	Inflammatory bowel disease 28
NM_001558.3(IL10RA):c.251C>T (p.Thr84Ile)	IL10RA	11	117989504	Inflammatory bowel disease 28
NM_001558.3(IL10RA):c.784C>T (p.Arg262Cys)	IL10RA	11	117995684	Inflammatory bowel disease 28
NM_001558.3(IL10RA):c.301C>T (p.Arg101Trp)	IL10RA	11	117989554	Inflammatory bowel disease 28
NM_005228.4(EGFR):c.1283G>A (p.Gly428Asp)	EGFR	7	55157738	Inflammatory skin and bowel disease, neonatal, 2
NM_001012339.2(DNAJC21):c.983+1G>A	DNAJC21	5	34941184	Inherited bone marrow failure syndrome
NM_000208.3(INSR):c.3540G>A (p.Met1180Ile)	INSR	19	7120739	Insulin resistance
NM_000208.3(INSR):c.3481G>A (p.Ala1161Thr)	INSR	19	7122662	Insulin resistance;insulin-resistant diabetes mellitus AND acanthosis nigricans
NM_014009.3(FOXP3):c.1189C>T (p.Arg397Trp)	FOXP3	X	49251441	Insulin-dependent diabetes mellitus secretory diarrhea syndrome
NM_014009.3(FOXP3):c.1150G>A (p.Ala384Thr)	FOXP3	X	49251480	Insulin-dependent diabetes mellitus secretory diarrhea syndrome
NM_014009.3(FOXP3):c.3G>A (p.Met1Ile)	FOXP3	X	49258503	Insulin-dependent diabetes mellitus secretory diarrhea syndrome
NM_000875.4(IGF1R):c.413G>A (p.Arg138Gln)	IGF1R	15	98707880	Insulin-like growth factor 1 resistance to
NM_000875.4(IGF1R):c.265C>T (p.Arg89Ter)	IGF1R	15	98707732	Insulin-like growth factor 1 resistance to
NM_000875.4(IGF1R):c.2216G>A (p.Arg739Gln)	IGF1R	15	98922162	Insulin-like growth factor 1 resistance to
NM_001111284.1(IGF1):c.226G>A (p.Val76Met)	IGF1	12	102419637	Insulin-like growth factor I deficiency
NM_001111283.2(IGF1):c.292C>T (p.Arg98Trp)	IGF1	12	102419619	Insulin-like growth factor I deficiency
NM_000208.3(INSR):c.479G>A (p.Trp160Ter)	INSR	19	7267518	Insulin-resistant diabetes mellitus AND acanthosis nigricans
NM_000208.3(INSR):c.3059G>A (p.Arg1020Gln)	INSR	19	7125482	Insulin-resistant diabetes mellitus AND acanthosis nigricans
NM_000208.3(INSR):c.3079C>T (p.Arg1027Ter)	INSR	19	7125462	Insulin-resistant diabetes mellitus AND acanthosis nigricans;Pineal hyperplasia AND diabetes mellitus syndrome
NM_001197104.1(KMT2A):c.3301C>T (p.Arg1101Ter)	KMT2A	11	118476949	Intellectual deficiency;not provided
NM_006578.3(GNB5):c.249G>A (p.Gln83=)	GNB5	15	52153940	INTELLECTUAL DEVELOPMENTAL DISORDER WITH CARDIAC ARRHYTHMIA
NM_006578.3(GNB5):c.994C>T (p.Arg332Ter)	GNB5	15	52124529	INTELLECTUAL DEVELOPMENTAL DISORDER WITH CARDIAC ARRHYTHMIA
NM_001003694.1(BRPF1):c.1108C>T (p.Pro370Ser)	BRPF1	3	9739507	INTELLECTUAL DEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES AND PTOSIS
NM_001003694.1(BRPF1):c.1363C>T (p.Arg455Ter)	BRPF1	3	9739762	INTELLECTUAL DEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES AND PTOSIS
NM_001003694.1(BRPF1):c.2497C>T (p.Arg833Ter)	BRPF1	3	9743763	INTELLECTUAL DEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES AND PTOSIS
NM_001003694.1(BRPF1):c.3298C>T (p.Arg1100Ter)	BRPF1	3	9745904	INTELLECTUAL DEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES AND PTOSIS
NM_018014.3(BCL11A):c.154C>T (p.Gln52Ter)	BCL11A	2	60546202	INTELLECTUAL DEVELOPMENTAL DISORDER WITH PERSISTENCE OF FETAL HEMOGLOBIN
NM_001083962.1(TCF4):c.520C>T (p.Arg174Ter)	TCF4	18	55350388	Intellectual disability
NM_001111125.2(IQSEC2):c.3097C>T (p.Gln1033Ter)	IQSEC2	X	53239213	Intellectual disability

NM_002024.5(FMR1):c.990+1G>A	FMR1	X	147936614	Intellectual disability
NM_001145715.2(KPNA7):c.106C>T (p.Arg36Ter)	KPNA7	7	99203201	Intellectual disability, mild
NM_005334.2(HCFC1):c.218C>T (p.Ala73Val)	HCFC1	X	153964702	Intellectual disability;Mental retardation 3, X-linked;not provided
NM_004992.3(MECP2):c.502C>T (p.Arg168Ter)	MECP2	X	154031326	Intellectual disability;Mental retardation, X-linked, syndromic 13;Rett syndrome;not provided
NM_004426.2(PHC1):c.2974C>T (p.Leu992Phe)	PHC1	12	8939418	Intellectual disability;Primary autosomal recessive microcephaly 11;Primary microcephaly
NM_000417.2(IL2RA):c.301C>T (p.Gln101Ter)	IL2RA	10	6024310	Interleukin 2 receptor, alpha, deficiency of
NM_000417.2(IL2RA):c.497G>A (p.Ser166Asn)	IL2RA	10	6021564	Interleukin 2 receptor, alpha, deficiency of
NM_002204.3(ITGA3):c.1387C>T (p.Arg463Trp)	ITGA3	17	50074452	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital
NM_002204.3(ITGA3):c.2070+1G>A	ITGA3	17	50077122	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital
NM_014967.4(FAN1):c.2245C>T (p.Arg749Ter)	FAN1	15	30925199	Interstitial nephritis, karyomegalic
NM_001110556.1(FLNA):c.5278C>T (p.Gln1760Ter)	FLNA	X	154354651	Intestinal pseudoobstruction neuronal chronic idiopathic X-linked;Periventricular nodular heterotopia 1
NM_000076.2(CDKN1C):c.820G>A (p.Asp274Asn)	CDKN1C	11	2884670	Intrauterine growth retardation, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomalies
NM_005142.2(GIF):c.137C>T (p.Ser46Leu)	GIF	11	59843998	Intrinsic factor deficiency
NM_203395.2(IYD):c.301C>T (p.Arg101Trp)	IYD	6	150389474	Iodotyrosine deiodination defect
NM_203395.2(IYD):c.658G>A (p.Ala220Thr)	IYD	6	150394226	Iodotyrosine deiodination defect
NM_003235.4(TG):c.4588C>T (p.Arg1530Ter)	TG	8	132923397	Iodotyrosyl coupling defect
NM_003235.4(TG):c.6725G>A (p.Arg2242His)	TG	8	133017940	Iodotyrosyl coupling defect
NM_003235.4(TG):c.7123G>A (p.Gly2375Arg)	TG	8	133029907	Iodotyrosyl coupling defect
NM_003235.4(TG):c.5690G>A (p.Cys1897Tyr)	TG	8	132967797	Iodotyrosyl coupling defect
NM_003235.4(TG):c.7007G>A (p.Arg2336Gln)	TG	8	133022121	Iodotyrosyl coupling defect
NM_003235.4(TG):c.6379C>T (p.Arg2127Ter)	TG	8	133012017	Iodotyrosyl coupling defect
NM_003235.4(TG):c.886C>T (p.Arg296Ter)	TG	8	132882609	Iodotyrosyl coupling defect;Thyroid dyshormonogenesis;not provided
NM_016123.3(IRAK4):c.877C>T (p.Gln293Ter)	IRAK4	12	43778238	IRAK4 deficiency
NM_016123.3(IRAK4):c.34C>T (p.Arg12Cys)	IRAK4	12	43768145	IRAK4 deficiency
NM_001453.2(FOXC1):c.392C>T (p.Ser131Leu)	FOXC1	6	1610837	Iridogoniodysgenesis type1
NM_153427.2(PITX2):c.250C>T (p.Arg84Trp)	PITX2	4	110621166	Iridogoniodysgenesis, dominant type
NM_153426.2(PITX2):c.344G>A (p.Arg115His)	PITX2	4	110621210	Iridogoniodysgenesis, dominant type;not provided
NM_003560.3(PLA2G6):c.2233C>T (p.Arg745Trp)	PLA2G6	22	38112547	Iron accumulation in brain
NM_003560.3(PLA2G6):c.2098C>T (p.Gln700Ter)	PLA2G6	22	38113591	Iron accumulation in brain
NM_003560.3(PLA2G6):c.1754C>T (p.Thr585Ile)	PLA2G6	22	38116200	Iron accumulation in brain
NM_003560.3(PLA2G6):c.1699G>A (p.Glu567Lys)	PLA2G6	22	38120802	Iron accumulation in brain
NM_003560.3(PLA2G6):c.1612C>T (p.Arg538Cys)	PLA2G6	22	38120889	Iron accumulation in brain
NM_003560.3(PLA2G6):c.517C>T (p.Gln173Ter)	PLA2G6	22	38143197	Iron accumulation in brain
NM_003560.3(PLA2G6):c.1903C>T (p.Arg635Ter)	PLA2G6	22	38115658	Iron accumulation in brain;not provided
NM_003560.3(PLA2G6):c.1799G>A (p.Arg600Gln)	PLA2G6	22	38116155	Iron accumulation in brain;not provided
NM_003560.3(PLA2G6):c.1613G>A (p.Arg538His)	PLA2G6	22	38120888	Iron accumulation in brain;not provided
NM_003560.3(PLA2G6):c.673C>T (p.His225Tyr)	PLA2G6	22	38140106	Iron accumulation in brain;not provided
NM_018488.3(TBX4):c.184C>T (p.Gln62Ter)	TBX4	17	61456674	Ischiopatellar dysplasia
NM_000525.3(KCNJ11):c.761C>T (p.Pro254Leu)	KCNJ11	11	17387331	Islet cell hyperplasia
NM_000525.3(KCNJ11):c.902G>A (p.Arg301His)	KCNJ11	11	17387190	Islet cell hyperplasia
NM_000525.3(KCNJ11):c.844G>A (p.Glu282Lys)	KCNJ11	11	17387248	Islet cell hyperplasia
NM_000525.3(KCNJ11):c.881C>T (p.Thr294Met)	KCNJ11	11	17387211	Islet cell hyperplasia
NM_000102.3(CYP17A1):c.1073G>A (p.Arg358Gln)	CYP17A1	10	102832577	Isolated 17,20-lyase deficiency

NM_000894.2(LHB):c.167G>A (p.Gly56Asp)	LHB	19	49016563	Isolated lutropin deficiency
NM_002225.3(IVD):c.157C>T (p.Arg53Cys)	IVD	15	40407639	Isovaleryl-CoA dehydrogenase deficiency
NM_002225.3(IVD):c.793+1G>A	IVD	15	40413088	Isovaleryl-CoA dehydrogenase deficiency
NM_002225.3(IVD):c.1117C>T (p.Gln373Ter)	IVD	15	40416332	Isovaleryl-CoA dehydrogenase deficiency
NM_002225.3(IVD):c.941C>T (p.Ala314Val)	IVD	15	40415454	Isovaleryl-CoA dehydrogenase deficiency;not provided
NM_002225.3(IVD):c.367G>A (p.Gly123Arg)	IVD	15	40410699	Isovaleryl-CoA dehydrogenase deficiency;not provided
NM_002225.3(IVD):c.241C>T (p.Arg81Ter)	IVD	15	40407723	Isovaleryl-CoA dehydrogenase deficiency;not provided
NM_004315.5(ASAH1):c.173C>T (p.Thr58Met)	ASAH1	8	18075541	Jankovic Rivera syndrome
NM_024753.4(TTC21B):c.684G>A (p.Trp228Ter)	TTC21B	2	165941053	Jeune thoracic dystrophy
NM_001080463.1(DYNC2H1):c.2702+1G>A	DYNC2H1	11	103143396	Jeune thoracic dystrophy
NM_001080463.1(DYNC2H1):c.10606C>T (p.Arg3536Ter)	DYNC2H1	11	103257731	Jeune thoracic dystrophy
NM_174916.2(UBR1):c.1537C>T (p.Gln513Ter)	UBR1	15	43048394	Johanson-Blizzard syndrome
NM_019892.5(INPP5E):c.1754G>A (p.Arg585His)	INPP5E	9	136430325	Joubert syndrome
NM_019892.5(INPP5E):c.1577C>T (p.Pro526Leu)	INPP5E	9	136431090	Joubert syndrome
NM_019892.5(INPP5E):c.1154G>A (p.Cys385Tyr)	INPP5E	9	136433160	Joubert syndrome
NM_019892.5(INPP5E):c.1064C>T (p.Thr355Met)	INPP5E	9	136433250	Joubert syndrome
NM_019892.5(INPP5E):c.1021G>A (p.Gly341Ser)	INPP5E	9	136434050	Joubert syndrome
NM_019892.5(INPP5E):c.944C>T (p.Pro315Leu)	INPP5E	9	136434127	Joubert syndrome
NM_019892.5(INPP5E):c.907G>A (p.Val303Met)	INPP5E	9	136434769	Joubert syndrome
NM_001286577.1(C2CD3):c.5267G>A (p.Gly1756Glu)	C2CD3	11	74049431	Joubert syndrome
NM_015681.4(B9D1):c.466C>T (p.Arg156Trp)	B9D1	17	19343796	Joubert syndrome
NM_001165927.1(MKS1):c.1231C>T (p.Pro411Ser)	MKS1	17	58207906	Joubert syndrome
NM_017777.3(MKS1):c.950G>A (p.Gly317Glu)	MKS1	17	58210988	Joubert syndrome
NM_030578.3(B9D2):c.463G>A (p.Gly155Ser)	B9D2	19	41354765	Joubert syndrome
NM_030578.3(B9D2):c.220C>T (p.Pro74Ser)	B9D2	19	41355008	Joubert syndrome
NM_001080522.2(CC2D2A):c.3652C>T (p.Arg1218Ter)	CC2D2A	4	15574207	Joubert syndrome
NM_019892.5(INPP5E):c.1543C>T (p.Arg515Trp)	INPP5E	9	136431830	Joubert syndrome 1
NM_019892.5(INPP5E):c.1688G>A (p.Arg563His)	INPP5E	9	136430391	Joubert syndrome 1
NM_019892.5(INPP5E):c.1132C>T (p.Arg378Cys)	INPP5E	9	136433182	Joubert syndrome 1
NM_003611.2(OFD1):c.2668C>T (p.Arg890Ter)	OFD1	X	13767195	Joubert syndrome 10;not provided
NM_018718.2(CEP41):c.536G>A (p.Arg179His)	CEP41	7	130402686	Joubert syndrome 12/15, digenic
NM_024549.5(TCTN1):c.856C>T (p.Arg286Ter)	TCTN1	12	110640437	Joubert syndrome 13
NM_001044385.2(TMEM237):c.76C>T (p.Gln26Ter)	TMEM237	2	201640264	Joubert syndrome 14
NM_016464.4(TMEM138):c.376G>A (p.Ala126Thr)	TMEM138	11	61367998	Joubert syndrome 16
NM_016464.4(TMEM138):c.380C>T (p.Ala127Val)	TMEM138	11	61368600	Joubert syndrome 16
NM_023073.3(C5orf42):c.7400+1G>A	C5orf42	5	37167046	Joubert syndrome 17
NM_023073.3(C5orf42):c.4006C>T (p.Arg1336Trp)	C5orf42	5	37187488	Joubert syndrome 17
NM_023073.3(C5orf42):c.4804C>T (p.Arg1602Ter)	C5orf42	5	37183377	Joubert syndrome 17
NM_023073.3(C5orf42):c.7477C>T (p.Arg2493Ter)	C5orf42	5	37165595	Joubert syndrome 17
NM_023073.3(C5orf42):c.8855+1G>A	C5orf42	5	37122429	Joubert syndrome 17
NM_023073.3(C5orf42):c.8710C>T (p.Arg2904Ter)	C5orf42	5	37125330	Joubert syndrome 17
NM_023073.3(C5orf42):c.8167C>T (p.Gln2723Ter)	C5orf42	5	37153784	Joubert syndrome 17
NM_023073.3(C5orf42):c.7978C>T (p.Arg2660Ter)	C5orf42	5	37153973	Joubert syndrome 17
NM_023073.3(C5orf42):c.7778G>A (p.Trp2593Ter)	C5orf42	5	37158258	Joubert syndrome 17

NM_023073.3(C5orf42):c.2923C>T (p.Gln975Ter)	C5orf42	5	37206423	Joubert syndrome 17
NM_023073.3(C5orf42):c.2831G>A (p.Arg944His)	C5orf42	5	37213648	Joubert syndrome 17
NM_023073.3(C5orf42):c.2709G>A (p.Trp903Ter)	C5orf42	5	37221361	Joubert syndrome 17
NM_023073.3(C5orf42):c.2611C>T (p.Arg871Cys)	C5orf42	5	37221459	Joubert syndrome 17
NM_023073.3(C5orf42):c.2377C>T (p.Gln793Ter)	C5orf42	5	37224655	Joubert syndrome 17
NM_023073.3(C5orf42):c.2275C>T (p.Gln759Ter)	C5orf42	5	37226320	Joubert syndrome 17
NM_023073.3(C5orf42):c.424G>A (p.Glu142Lys)	C5orf42	5	37244521	Joubert syndrome 17
NM_023073.3(C5orf42):c.9058C>T (p.Arg3020Ter)	C5orf42	5	37120306	Joubert syndrome 17;not provided
NM_023073.3(C5orf42):c.3599C>T (p.Ala1200Val)	C5orf42	5	37198775	Joubert syndrome 17;not provided
NM_023073.3(C5orf42):c.2353C>T (p.Arg785Ter)	C5orf42	5	37224679	Joubert syndrome 17;not provided
NM_015631.5(TCTN3):c.940G>A (p.Gly314Arg)	TCTN3	10	95685585	Joubert syndrome 18
NM_015631.5(TCTN3):c.3G>A (p.Met1Ile)	TCTN3	10	95693897	Joubert syndrome 18
NM_001173990.2(TMEM216):c.217C>T (p.Arg73Cys)	TMEM216	11	61393964	Joubert syndrome 2
NM_001173990.2(TMEM216):c.218G>A (p.Arg73His)	TMEM216	11	61393965	Joubert syndrome 2;Meckel syndrome type 2
NM_001173990.2(TMEM216):c.253C>T (p.Arg85Ter)	TMEM216	11	61397797	Joubert syndrome 2;Meckel syndrome type 2
NM_001077416.2(TMEM231):c.784G>A (p.Asp262Asn)	TMEM231	16	75542641	Joubert syndrome 20;not provided
NM_024790.6(CSPP1):c.2320C>T (p.Arg774Ter)	CSPP1	8	67158540	Joubert syndrome 21
NM_024790.6(CSPP1):c.2953+1G>A	CSPP1	8	67172556	Joubert syndrome 21
NM_024790.6(CSPP1):c.658C>T (p.Arg220Ter)	CSPP1	8	67095440	Joubert syndrome 21
NM_024790.6(CSPP1):c.652C>T (p.Gln218Ter)	CSPP1	8	67095434	Joubert syndrome 21
NM_024790.6(CSPP1):c.2773C>T (p.Arg925Ter)	CSPP1	8	67164468	Joubert syndrome 21
NM_024790.6(CSPP1):c.1132C>T (p.Arg378Ter)	CSPP1	8	67111983	Joubert syndrome 21
NM_024790.6(CSPP1):c.2260C>T (p.Arg754Ter)	CSPP1	8	67158480	Joubert syndrome 21
NM_024790.6(CSPP1):c.3205+1G>A	CSPP1	8	67179927	Joubert syndrome 21
NM_002601.3(PDE6D):c.140-1G>A	PDE6D	2	231738139	Joubert syndrome 22
NM_001244189.1(KIAA0586):c.1159C>T (p.Gln387Ter)	KIAA0586	14	58450617	Joubert syndrome 23
NM_001244189.1(KIAA0586):c.802C>T (p.Gln268Ter)	KIAA0586	14	58444011	Joubert syndrome 23
NM_001244189.1(KIAA0586):c.3303G>A (p.Pro1101=)	KIAA0586	14	58482712	Joubert syndrome 23
NM_001244189.1(KIAA0586):c.949C>T (p.Gln317Ter)	KIAA0586	14	58444158	Joubert syndrome 23
NM_024809.4(TCTN2):c.1235_1312del (p.Ile413_Gly438del)	TCTN2	12	123695219	Joubert syndrome 24
NM_024809.4(TCTN2):c.1873C>T (p.Gln625Ter)	TCTN2	12	123706829	Joubert syndrome 24
NM_014704.3(CEP104):c.496C>T (p.Arg166Ter)	CEP104	1	3844977	Joubert syndrome 25
NM_001243473.2(B9D1):c.526G>A (p.Gly176Arg)	B9D1	17	19343795	JOUBERT SYNDROME 27
NM_001134831.1(AHI1):c.1051C>T (p.Arg351Ter)	AHI1	6	135457594	Joubert syndrome 3
NM_001134831.1(AHI1):c.1303C>T (p.Arg435Ter)	AHI1	6	135455775	Joubert syndrome 3
NM_001134831.1(AHI1):c.1765C>T (p.Arg589Ter)	AHI1	6	135447022	Joubert syndrome 3
NM_001134831.1(AHI1):c.1484G>A (p.Arg495His)	AHI1	6	135448432	Joubert syndrome 3
NM_017651.4(AHI1):c.2174G>A (p.Trp725Ter)	AHI1	6	135433119	Joubert syndrome 3
NM_001134831.1(AHI1):c.2297G>A (p.Gly766Glu)	AHI1	6	135431284	Joubert syndrome 3
NM_001134831.1(AHI1):c.2023G>A (p.Asp675Asn)	AHI1	6	135438388	Joubert syndrome 3
NM_001134831.1(AHI1):c.2012C>T (p.Thr671Ile)	AHI1	6	135438399	Joubert syndrome 3
NM_001134831.1(AHI1):c.1626+1G>A	AHI1	6	135448289	Joubert syndrome 3
NM_001134831.1(AHI1):c.1516C>T (p.Arg506Ter)	AHI1	6	135448400	Joubert syndrome 3
NM_001134831.1(AHI1):c.1260G>A (p.Trp420Ter)	AHI1	6	135455818	Joubert syndrome 3

NM_017651.4(AHI1):c.985C>T (p.Arg329Ter)	AHI1	6	135457660	Joubert syndrome 3;not provided
NM_001134831.1(AHI1):c.2212C>T (p.Arg738Ter)	AHI1	6	135433081	Joubert syndrome 3;not provided
NM_025114.3(CEP290):c.5932C>T (p.Arg1978Ter)	CEP290	12	88071373	Joubert syndrome 5
NM_025114.3(CEP290):c.5344C>T (p.Arg1782Ter)	CEP290	12	88079112	Joubert syndrome 5
NM_025114.3(CEP290):c.4522C>T (p.Arg1508Ter)	CEP290	12	88084768	Joubert syndrome 5
NM_025114.3(CEP290):c.4393C>T (p.Arg1465Ter)	CEP290	12	88086083	Joubert syndrome 5
NM_025114.3(CEP290):c.1623+1G>A	CEP290	12	88118642	Joubert syndrome 5
NM_025114.3(CEP290):c.2722C>T (p.Arg908Ter)	CEP290	12	88106770	Joubert syndrome 5
NM_025114.3(CEP290):c.2668C>T (p.Gln890Ter)	CEP290	12	88106824	Joubert syndrome 5
NM_025114.3(CEP290):c.3904C>T (p.Gln1302Ter)	CEP290	12	88089157	Joubert syndrome 5;Leber congenital amaurosis 10
NM_025114.3(CEP290):c.1984C>T (p.Gln662Ter)	CEP290	12	88114488	Joubert syndrome 5;Meckel syndrome type 4
NM_001142301.1(TMEM67):c.1391G>A (p.Gly464Glu)	TMEM67	8	93793256	Joubert syndrome 6
NM_153704.5(TMEM67):c.130C>T (p.Gln44Ter)	TMEM67	8	93755044	Joubert syndrome 6
NM_153704.5(TMEM67):c.244C>T (p.Pro82Ser)	TMEM67	8	93755798	Joubert syndrome 6
NM_153704.5(TMEM67):c.515G>A (p.Arg172Gln)	TMEM67	8	93765414	Joubert syndrome 6
NM_153704.5(TMEM67):c.1073C>T (p.Pro358Leu)	TMEM67	8	93782402	Joubert syndrome 6
NM_153704.5(TMEM67):c.1351C>T (p.Arg451Ter)	TMEM67	8	93786285	Joubert syndrome 6
NM_153704.5(TMEM67):c.1453C>T (p.Pro485Ser)	TMEM67	8	93787884	Joubert syndrome 6
NM_153704.5(TMEM67):c.2290C>T (p.Arg764Ter)	TMEM67	8	93803652	Joubert syndrome 6
NM_153704.5(TMEM67):c.2801G>A (p.Gly934Glu)	TMEM67	8	93815341	Joubert syndrome 6
NM_153704.5(TMEM67):c.675G>A (p.Trp225Ter)	TMEM67	8	93772612	Joubert syndrome 6;Meckel syndrome type 3
NM_001127897.3(RPGRIP1L):c.757C>T (p.Gln253Ter)	RPGRIP1L	16	53686452	Joubert syndrome 7
NM_015272.4(RPGRIP1L):c.2050C>T (p.Gln684Ter)	RPGRIP1L	16	53652637	Joubert syndrome 7
NM_015272.4(RPGRIP1L):c.3529C>T (p.Arg1177Ter)	RPGRIP1L	16	53619112	Joubert syndrome 7
NM_015272.4(RPGRIP1L):c.2305-1G>A	RPGRIP1L	16	53646004	Joubert syndrome 7
NM_015272.4(RPGRIP1L):c.1243+1G>A	RPGRIP1L	16	53664869	Joubert syndrome 7
NM_001127897.3(RPGRIP1L):c.2030C>T (p.Thr677Ile)	RPGRIP1L	16	53652657	Joubert syndrome 7;Meckel syndrome type 5
NM_001127897.3(RPGRIP1L):c.2614C>T (p.Gln872Ter)	RPGRIP1L	16	53645694	Joubert syndrome 7;Meckel syndrome type 5;not provided
NM_182896.2(ARL13B):c.236G>A (p.Arg79Gln)	ARL13B	3	94003764	Joubert syndrome 8
NM_182896.2(ARL13B):c.246G>A (p.Trp82Ter)	ARL13B	3	94003774	Joubert syndrome 8
NM_182896.2(ARL13B):c.598C>T (p.Arg200Cys)	ARL13B	3	94036663	Joubert syndrome 8
NM_001080522.2(CC2D2A):c.3364C>T (p.Pro1122Ser)	CC2D2A	4	15567752	Joubert syndrome 9
NM_001080522.2(CC2D2A):c.2848C>T (p.Arg950Ter)	CC2D2A	4	15559183	Joubert syndrome 9
NM_001080522.2(CC2D2A):c.2683C>T (p.Gln895Ter)	CC2D2A	4	15557361	Joubert syndrome 9
NM_001080522.2(CC2D2A):c.2671G>A (p.Glu891Lys)	CC2D2A	4	15557349	Joubert syndrome 9
NM_001080522.2(CC2D2A):c.3055C>T (p.Arg1019Ter)	CC2D2A	4	15563395	Joubert syndrome 9
NM_001080522.2(CC2D2A):c.3850C>T (p.Arg1284Cys)	CC2D2A	4	15580046	Joubert syndrome 9
NM_001080522.2(CC2D2A):c.3989G>A (p.Arg1330Gln)	CC2D2A	4	15586170	Joubert syndrome 9
NM_018718.2(CEP41):c.1078C>T (p.Arg360Cys)	CEP41	7	130398935	Joubert syndrome 9/15, digenic
NM_001080522.2(CC2D2A):c.3341C>T (p.Thr1114Met)	CC2D2A	4	15567729	Joubert syndrome 9;Meckel syndrome type 6
NM_001080522.2(CC2D2A):c.4179+1G>A	CC2D2A	4	15587930	Joubert syndrome 9;Meckel syndrome type 6
NM_001080522.2(CC2D2A):c.4582C>T (p.Arg1528Cys)	CC2D2A	4	15599614	Joubert syndrome 9;not provided
NM_025114.3(CEP290):c.4811G>A (p.Trp1604Ter)	CEP290	12	88083848	Joubert Syndrome and Related Disorders
NM_019892.5(INPP5E):c.1304G>A (p.Arg435Gln)	INPP5E	9	136432562	Joubert syndrome;Joubert syndrome 1
NM_001044385.2(TMEM237):c.52C>T (p.Arg18Ter)	TMEM237	2	201640915	Joubert syndrome;Joubert syndrome 14

NM_015202.3(KIAA0556):c.2674C>T (p.Gln892Ter)	KIAA0556	16	27749634	Joubert syndrome;Joubert syndrome 26
NM_001134831.1(AHI1):c.2168G>A (p.Arg723Gln)	AHI1	6	135433125	Joubert syndrome;Joubert syndrome 3
NM_001134831.1(AHI1):c.1267C>T (p.Gln423Ter)	AHI1	6	135455811	Joubert syndrome;Joubert syndrome 3;not provided
NM_001080522.2(CC2D2A):c.1558C>T (p.Arg520Ter)	CC2D2A	4	15533284	Joubert syndrome;Joubert syndrome 9;Meckel-Gruber syndrome
NM_001080522.2(CC2D2A):c.1017+1G>A	CC2D2A	4	15516005	Joubert syndrome;Joubert syndrome 9;not provided
NM_025114.3(CEP290):c.4437+1G>A	CEP290	12	88086038	Joubert syndrome;Leber congenital amaurosis 10;Meckel-Gruber syndrome;Nephronophthisis
NM_001080522.2(CC2D2A):c.1267C>T (p.Arg423Ter)	CC2D2A	4	15527564	Joubert syndrome;Meckel-Gruber syndrome
NM_017777.3(MKS1):c.1208C>T (p.Ser403Leu)	MKS1	17	58207959	Joubert syndrome;not provided
NM_003611.2(OFD1):c.2725C>T (p.Arg909Ter)	OFD1	X	13767252	Joubert syndrome;Oral-facial-digital syndrome
NM_001286577.1(C2CD3):c.184C>T (p.Arg62Ter)	C2CD3	11	74168485	Joubert syndrome;Orofaciodigital syndrome xiv
NM_015662.2(IFT172):c.4630C>T (p.Arg1544Cys)	IFT172	2	27447544	Joubert syndrome;Short-rib thoracic dysplasia 10 with or without polydactyly
NM_198129.2(LAMA3):c.6808C>T (p.Arg2270Ter)	LAMA3	18	23907639	Junctional epidermolysis bullosa gravis of Herlitz
NM_000228.2(LAMB3):c.496C>T (p.Gln166Ter)	LAMB3	1	209634515	Junctional epidermolysis bullosa gravis of Herlitz
NM_000228.2(LAMB3):c.1830G>A (p.Trp610Ter)	LAMB3	1	209625794	Junctional epidermolysis bullosa gravis of Herlitz
NM_000228.2(LAMB3):c.2806C>T (p.Gln936Ter)	LAMB3	1	209618555	Junctional epidermolysis bullosa gravis of Herlitz
NM_005562.2(LAMC2):c.283C>T (p.Arg95Ter)	LAMC2	1	183215467	Junctional epidermolysis bullosa gravis of Herlitz
NM_000228.2(LAMB3):c.727C>T (p.Gln243Ter)	LAMB3	1	209632678	Junctional epidermolysis bullosa gravis of Herlitz
NM_000228.2(LAMB3):c.1117C>T (p.Gln373Ter)	LAMB3	1	209629752	Junctional epidermolysis bullosa gravis of Herlitz
NM_000228.2(LAMB3):c.124C>T (p.Arg42Ter)	LAMB3	1	209650023	Junctional epidermolysis bullosa gravis of Herlitz;not provided
NM_000228.2(LAMB3):c.1705C>T (p.Arg569Ter)	LAMB3	1	209625919	Junctional epidermolysis bullosa gravis of Herlitz;not provided
NM_001793.5(CDH3):c.1508G>A (p.Arg503His)	CDH3	16	68685288	Juvenile macular degeneration and hypotrichosis
NM_001793.5(CDH3):c.1085G>A (p.Trp362Ter)	CDH3	16	68682390	Juvenile macular degeneration and hypotrichosis
NM_000267.3(NF1):c.4614G>A (p.Trp1538Ter)	NF1	17	31261810	Juvenile myelomonocytic leukemia
NM_018100.3(EFHC1):c.628G>A (p.Asp210Asn)	EFHC1	6	52452742	Juvenile myoclonic epilepsy
m.583G>A	MT-TF	MT	583	Juvenile myopathy, encephalopathy, lactic acidosis AND stroke
m.4332G>A	MT-TQ	MT	4332	Juvenile myopathy, encephalopathy, lactic acidosis AND stroke
m.14453G>A	MT-ND6	MT	14453	Juvenile myopathy, encephalopathy, lactic acidosis AND stroke
m.3946G>A	MT-ND1	MT	3946	Juvenile myopathy, encephalopathy, lactic acidosis AND stroke
m.3697G>A	MT-ND1	MT	3697	Juvenile myopathy, encephalopathy, lactic acidosis AND stroke;Leber hereditary optic neuropathy with dystonia;Leber's optic atrophy
m.13042G>A	MT-ND5	MT	13042	Juvenile myopathy, encephalopathy, lactic acidosis AND stroke;Leigh syndrome due to mitochondrial complex I deficiency;Myoclonus with epilepsy with ragged red fibers
m.13513G>A	MT-ND5;MT-	MT	13513	Juvenile myopathy, encephalopathy, lactic acidosis AND stroke;Leigh syndrome;Leigh syndrome due to mitochondrial complex I deficiency;not provided
NM_001256214.1(ATP1A3):c.424G>A (p.Val142Met)	ATP1A3	19	41986202	Juvenile onset psychosis
NM_004329.2(BMPR1A):c.715C>T (p.Gln239Ter)	BMPR1A	10	86917173	Juvenile polyposis syndrome
NM_004329.2(BMPR1A):c.812G>A (p.Trp271Ter)	BMPR1A	10	86917270	Juvenile polyposis syndrome
NM_004329.2(BMPR1A):c.1127G>A (p.Cys376Tyr)	BMPR1A	10	86919430	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.1162C>T (p.Gln388Ter)	SMAD4	18	51067041	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.403C>T (p.Arg135Ter)	SMAD4	18	51048839	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.538C>T (p.Gln180Ter)	SMAD4	18	51054864	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.1139+1G>A	SMAD4	18	51065607	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.1168G>A (p.Glu390Lys)	SMAD4	18	51067047	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.1193G>A (p.Trp398Ter)	SMAD4	18	51067072	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.1342C>T (p.Gln448Ter)	SMAD4	18	51076671	Juvenile polyposis syndrome

NM_005359.5(SMAD4):c.1527G>A (p.Trp509Ter)	SMAD4	18	51078335	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.1139G>A (p.Arg380Lys)	SMAD4	18	51065606	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.424+1G>A	SMAD4	18	51048861	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.906G>A (p.Trp302Ter)	SMAD4	18	51059867	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.1096C>T (p.Gln366Ter)	SMAD4	18	51065563	Juvenile polyposis syndrome
NM_005359.5(SMAD4):c.302G>A (p.Trp101Ter)	SMAD4	18	51048738	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
NM_005359.5(SMAD4):c.1055G>A (p.Gly352Glu)	SMAD4	18	51065522	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
NM_005359.5(SMAD4):c.1600C>T (p.Gln534Ter)	SMAD4	18	51078408	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
NM_005359.5(SMAD4):c.1447+1G>A	SMAD4	18	51076777	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
NM_005359.5(SMAD4):c.1054G>A (p.Gly352Arg)	SMAD4	18	51065521	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;not provided
NM_020919.3(ALS2):c.1619G>A (p.Gly540Glu)	ALS2	2	201754524	Juvenile primary lateral sclerosis
NM_000330.3(RS1):c.214G>A (p.Glu72Lys)	RS1	X	18647303	Juvenile retinoschisis;not provided
NM_000330.3(RS1):c.608C>T (p.Pro203Leu)	RS1	X	18642071	Juvenile retinoschisis;not provided
NM_000330.3(RS1):c.305G>A (p.Arg102Gln)	RS1	X	18647212	Juvenile retinoschisis;not provided
NM_000330.3(RS1):c.422G>A (p.Arg141His)	RS1	X	18644530	Juvenile retinoschisis;not provided
NM_000330.3(RS1):c.522+1G>A	RS1	X	18644429	Juvenile retinoschisis;not provided
NM_000330.3(RS1):c.304C>T (p.Arg102Trp)	RS1	X	18647213	Juvenile retinoschisis;Retinal dystrophy;not provided
NM_001101.3(ACTB):c.547C>T (p.Arg183Trp)	ACTB	7	5528536	Juvenile-onset dystonia
NM_003482.3(KMT2D):c.15536G>A (p.Arg5179His)	KMT2D	12	49026430	Kabuki syndrome 1
NM_003482.3(KMT2D):c.16360C>T (p.Arg5454Ter)	KMT2D	12	49022332	Kabuki syndrome 1
NM_003482.3(KMT2D):c.15943C>T (p.Gln5315Ter)	KMT2D	12	49024687	Kabuki syndrome 1
NM_003482.3(KMT2D):c.15844C>T (p.Arg5282Ter)	KMT2D	12	49024887	Kabuki syndrome 1
NM_003482.3(KMT2D):c.15791G>A (p.Trp5264Ter)	KMT2D	12	49024940	Kabuki syndrome 1
NM_003482.3(KMT2D):c.15195G>A (p.Trp5065Ter)	KMT2D	12	49026771	Kabuki syndrome 1
NM_003482.3(KMT2D):c.13606C>T (p.Arg4536Ter)	KMT2D	12	49030958	Kabuki syndrome 1
NM_003482.3(KMT2D):c.13450C>T (p.Arg4484Ter)	KMT2D	12	49031255	Kabuki syndrome 1
NM_003482.3(KMT2D):c.12592C>T (p.Arg4198Ter)	KMT2D	12	49032113	Kabuki syndrome 1
NM_003482.3(KMT2D):c.11290C>T (p.Gln3764Ter)	KMT2D	12	49033415	Kabuki syndrome 1
NM_003482.3(KMT2D):c.11263C>T (p.Gln3755Ter)	KMT2D	12	49033442	Kabuki syndrome 1
NM_003482.3(KMT2D):c.8743C>T (p.Arg2915Ter)	KMT2D	12	49038613	Kabuki syndrome 1
NM_003482.3(KMT2D):c.8053C>T (p.Arg2685Ter)	KMT2D	12	49039611	Kabuki syndrome 1
NM_003482.3(KMT2D):c.3553C>T (p.Gln1185Ter)	KMT2D	12	49050035	Kabuki syndrome 1
NM_003482.3(KMT2D):c.3121C>T (p.Gln1041Ter)	KMT2D	12	49050467	Kabuki syndrome 1
NM_003482.3(KMT2D):c.4265G>A (p.Trp1422Ter)	KMT2D	12	49046762	Kabuki syndrome 1
NM_003482.3(KMT2D):c.5677C>T (p.Gln1893Ter)	KMT2D	12	49042846	Kabuki syndrome 1
NM_003482.3(KMT2D):c.7903C>T (p.Arg2635Ter)	KMT2D	12	49039867	Kabuki syndrome 1
NM_003482.3(KMT2D):c.11845C>T (p.Gln3949Ter)	KMT2D	12	49032860	Kabuki syndrome 1
NM_003482.3(KMT2D):c.11275C>T (p.Gln3759Ter)	KMT2D	12	49033430	Kabuki syndrome 1
NM_003482.3(KMT2D):c.11770C>T (p.Gln3924Ter)	KMT2D	12	49032935	Kabuki syndrome 1
NM_003482.3(KMT2D):c.11713C>T (p.Gln3905Ter)	KMT2D	12	49032992	Kabuki syndrome 1
NM_003482.3(KMT2D):c.5104C>T (p.Arg1702Ter)	KMT2D	12	49044284	Kabuki syndrome 1
NM_003482.3(KMT2D):c.14075+1G>A	KMT2D	12	49029400	Kabuki syndrome 1
NM_003482.3(KMT2D):c.12661C>T (p.Gln4221Ter)	KMT2D	12	49032044	Kabuki syndrome 1
NM_003482.3(KMT2D):c.10810C>T (p.Gln3604Ter)	KMT2D	12	49033895	Kabuki syndrome 1
NM_003482.3(KMT2D):c.10819C>T (p.Gln3607Ter)	KMT2D	12	49033886	Kabuki syndrome 1;not provided

NM_003482.3(KMT2D):c.10834C>T (p.Gln3612Ter)	KMT2D	12	49033871	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.11149C>T (p.Gln3717Ter)	KMT2D	12	49033556	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.11692C>T (p.Gln3898Ter)	KMT2D	12	49033013	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.12406C>T (p.Gln4136Ter)	KMT2D	12	49032299	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.12430C>T (p.Gln4144Ter)	KMT2D	12	49032275	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.14710C>T (p.Arg4904Ter)	KMT2D	12	49027256	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.3532C>T (p.Gln1178Ter)	KMT2D	12	49050056	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.7066C>T (p.Gln2356Ter)	KMT2D	12	49040704	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.15061C>T (p.Arg5021Ter)	KMT2D	12	49026965	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.15256C>T (p.Arg5086Ter)	KMT2D	12	49026710	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.3754C>T (p.Arg1252Ter)	KMT2D	12	49049834	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.8488C>T (p.Arg2830Ter)	KMT2D	12	49038868	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.2797+1G>A	KMT2D	12	49050885	Kabuki syndrome 1;not provided
NM_003482.3(KMT2D):c.15079C>T (p.Arg5027Ter)	KMT2D	12	49026887	Kabuki syndrome 1;not provided
NM_001291415.1(KDM6A):c.3873G>A (p.Trp1291Ter)	KDM6A	X	45089911	Kabuki syndrome 2
NM_001291415.1(KDM6A):c.1711C>T (p.Arg571Ter)	KDM6A	X	45063449	Kabuki syndrome 2
NM_000216.3(ANOS1):c.711G>A (p.Trp237Ter)	ANOS1	X	8587809	Kallmann syndrome 1
NM_000216.3(ANOS1):c.769C>T (p.Arg257Ter)	ANOS1	X	8585354	Kallmann syndrome 1
NM_000216.3(ANOS1):c.774G>A (p.Trp258Ter)	ANOS1	X	8585349	Kallmann syndrome 1
NM_000216.3(ANOS1):c.1540G>A (p.Glu514Lys)	ANOS1	X	8536852	Kallmann syndrome 1
NM_000216.3(ANOS1):c.784C>T (p.Arg262Ter)	ANOS1	X	8585339	Kallmann syndrome 1
NM_000216.3(ANOS1):c.1187C>T (p.Ser396Leu)	ANOS1	X	8568252	Kallmann syndrome 1
NM_023110.2(FGFR1):c.709G>A (p.Gly237Ser)	FGFR1	8	38426158	Kallmann syndrome 2
NM_023110.2(FGFR1):c.142G>A (p.Gly48Ser)	FGFR1	8	38429898	Kallmann syndrome 2
NM_023110.2(FGFR1):c.749G>A (p.Arg250Gln)	FGFR1	8	38424696	Kallmann syndrome 2
NM_023110.2(FGFR1):c.443G>A (p.Arg148His)	FGFR1	8	38428351	Kallmann syndrome 2
NM_023110.2(FGFR1):c.1460G>A (p.Gly487Asp)	FGFR1	8	38417962	Kallmann syndrome 2
NM_023110.2(FGFR1):c.2084C>T (p.Thr695Ile)	FGFR1	8	38414254	Kallmann syndrome 2
NM_023110.2(FGFR1):c.565C>T (p.Arg189Cys)	FGFR1	8	38427977	Kallmann syndrome 2
NM_144773.3(PROKR2):c.969G>A (p.Met323Ile)	PROKR2	20	5302226	Kallmann syndrome 3
NM_144773.3(PROKR2):c.343G>A (p.Val115Met)	PROKR2	20	5314027	Kallmann syndrome 3
NM_001126128.1(PROK2):c.217C>T (p.Arg73Cys)	PROK2	3	71781472	Kallmann syndrome 4
NM_001126128.1(PROK2):c.101G>A (p.Cys34Tyr)	PROK2	3	71781588	Kallmann syndrome 4
NM_000262.2(NAGA):c.985C>T (p.Arg329Trp)	NAGA	22	42061040	Kanzaki disease
NM_000262.2(NAGA):c.986G>A (p.Arg329Gln)	NAGA	22	42061039	Kanzaki disease
NM_003560.3(PLA2G6):c.1894C>T (p.Arg632Trp)	PLA2G6	22	38115667	Karak syndrome
NM_012144.3(DNAI1):c.1490G>A (p.Gly497Asp)	DNAI1	9	34513112	Kartagener syndrome
NM_012144.3(DNAI1):c.2001+1G>A (p.Ala607_Lys667del)	DNAI1	9	34517468	Kartagener syndrome
NM_001190787.1(MCIDAS):c.1142G>A (p.Arg381His)	MCIDAS	5	55220382	Kartagener syndrome
NM_001190787.1(MCIDAS):c.1097G>A (p.Gly366Asp)	MCIDAS	5	55220427	Kartagener syndrome
NM_080860.3(RSPH1):c.727+5G>A (p.Ala244ValfsTer22)	RSPH1	21	42477286	Kartagener syndrome
NM_080860.3(RSPH1):c.366G>A (p.Arg122=)	RSPH1	21	42485804	Kartagener syndrome
NM_012144.3(DNAI1):c.1543G>A (p.Gly515Ser)	DNAI1	9	34513165	Kartagener syndrome;Primary ciliary dyskinesia

NM_006766.4(KAT6A):c.3385C>T (p.Arg1129Ter)	KAT6A	8	41934835	KAT6A syndrome;Mental retardation, autosomal dominant 32
NM_006766.4(KAT6A):c.3070C>T (p.Arg1024Ter)	KAT6A	8	41937538	KAT6A syndrome;Mental retardation, autosomal dominant 32
NM_001256182.1(ANKRD11):c.7180C>T (p.Gln2394Ter)	ANKRD11	16	89279362	KBG syndrome
NM_013275.5(ANKRD11):c.2197C>T (p.Arg733Ter)	ANKRD11	16	89284345	KBG syndrome;not provided
m.5877C>T	MT-TY	MT	5877	Kearns Sayre syndrome
m.3249G>A	MT-TL1	MT	3249	Kearns Sayre syndrome
NM_002240.4(KCNJ6):c.460G>A (p.Gly154Ser)	KCNJ6	21	37714697	Keppen-Lubinsky syndrome
NM_004004.5(GJB2):c.50C>T (p.Ser17Phe)	GJB2	13	20189532	Keratitis-ichthyosis-deafness syndrome, autosomal dominant
NM_014588.5(VSX1):c.496C>T (p.Arg166Trp)	VSX1	20	25079443	Keratoconus 1
NM_001127464.2(ZNF469):c.11101G>A (p.Gly3701Ser)	ZNF469	16	88438655	Keratoconus 1
NM_001127464.2(ZNF469):c.11615C>T (p.Pro3872Leu)	ZNF469	16	88439169	Keratoconus 1
NM_001127464.2(ZNF469):c.290C>T (p.Pro97Leu)	ZNF469	16	88427760	Keratoconus 1
NM_001127464.2(ZNF469):c.3376G>A (p.Glu1113Lys)	ZNF469	16	88427807	Keratoconus 1
NM_001681.3(ATP2A2):c.322C>T (p.Gln108Ter)	ATP2A2	12	110292122	Keratosis follicularis
NM_001681.3(ATP2A2):c.392G>A (p.Arg131Gln)	ATP2A2	12	110296666	Keratosis follicularis
NM_170665.3(ATP2A2):c.68G>A (p.Gly23Glu)	ATP2A2	12	110281857	Keratosis follicularis;not provided
NM_001942.3(DSG1):c.76C>T (p.Arg26Ter)	DSG1	18	31326608	Keratosis palmoplantaris striata 1
NM_001942.3(DSG1):c.601C>T (p.Gln201Ter)	DSG1	18	31331784	Keratosis palmoplantaris striata 1
NM_004415.3(DSP):c.991C>T (p.Gln331Ter)	DSP	6	7566428	Keratosis palmoplantaris striata II
NM_001190839.2(MGP):c.169+1G>A	MGP	12	14884212	Keutel syndrome
NM_017671.4(FERMT1):c.787C>T (p.Gln263Ter)	FERMT1	20	6107594	Kindler's syndrome
NM_017671.4(FERMT1):c.811C>T (p.Arg271Ter)	FERMT1	20	6107570	Kindler's syndrome
NM_017671.4(FERMT1):c.862C>T (p.Arg288Ter)	FERMT1	20	6097619	Kindler's syndrome
NM_017671.4(FERMT1):c.1848G>A (p.Trp616Ter)	FERMT1	20	6079448	Kindler's syndrome
NM_017671.4(FERMT1):c.1139+740G>A	FERMT1	20	6094199	Kindler's syndrome
NM_017671.4(FERMT1):c.958-1G>A	FERMT1	20	6097034	Kindler's syndrome
NM_017671.4(FERMT1):c.957+1G>A	FERMT1	20	6097523	Kindler's syndrome
NM_000893.3(KNG1):c.586C>T (p.Arg196Ter)	KNG1	3	186727258	Kininogen deficiency, total
NM_001127366.2(PAX3):c.251C>T (p.Ser84Phe)	PAX3	2	222297048	Klein-Waardenberg's syndrome;Waardenburg syndrome type 1
NM_004527.3(MEOX1):c.250C>T (p.Gln84Ter)	MEOX1	17	43661285	Klippel-Feil syndrome 2, autosomal recessive
NM_001844.4(COL2A1):c.1023+1G>A	COL2A1	12	47992877	Kniest dysplasia
NM_001844.4(COL2A1):c.908G>A (p.Gly303Asp)	COL2A1	12	47993825	Kniest dysplasia;Stickler syndrome type 1
NM_004004.5(GJB2):c.175G>A (p.Gly59Ser)	GJB2	13	20189407	Knuckle pads, deafness AND leukonychia syndrome
NM_024589.2(ROGDI):c.286C>T (p.Gln96Ter)	ROGDI	16	4800548	Kohlschutter's syndrome
NM_024589.2(ROGDI):c.469C>T (p.Arg157Ter)	ROGDI	16	4798631	Kohlschutter's syndrome
NM_001193466.1(KANSL1):c.1816C>T (p.Arg606Ter)	KANSL1	17	46066569	Koolen-de Vries syndrome
NM_001193466.1(KANSL1):c.916C>T (p.Gln306Ter)	KANSL1	17	46171228	Koolen-de Vries syndrome
NM_001193466.1(KANSL1):c.1652+1G>A	KANSL1	17	46067548	Koolen-de Vries syndrome;not provided
NM_000515.4(GH1):c.307C>T (p.Arg103Cys)	GH1	17	63917909	Kowarski syndrome
NM_000344.3(SMN1):c.821C>T (p.Thr274Ile)	SMN1	5	70946163	Kugelberg-Welander disease;Spinal muscular atrophy, type II
NM_000344.3(SMN1):c.305G>A (p.Trp102Ter)	SMN1	5	70942389	Kugelberg-Welander disease;Spinal muscular atrophy, type II
NM_024884.2(L2HGDH):c.905C>T (p.Pro302Leu)	L2HGDH	14	50269164	L-2-hydroxyglutaric aciduria
NM_024884.2(L2HGDH):c.164G>A (p.Gly55Asp)	L2HGDH	14	50302994	L-2-hydroxyglutaric aciduria
NM_024884.2(L2HGDH):c.1003C>T (p.Arg335Ter)	L2HGDH	14	50267814	L-2-hydroxyglutaric aciduria
NM_005670.3(EPM2A):c.835G>A (p.Gly279Ser)	EPM2A	6	145627577	Lafora disease

NM_005670.3(EPM2A):c.322C>T (p.Arg108Cys)	EPM2A	6	145686276	Lafora disease
NM_005670.3(EPM2A):c.721C>T (p.Arg241Ter)	EPM2A	6	145627691	Lafora disease;not provided
NM_198586.2(NHLRC1):c.436G>A (p.Asp146Asn)	NHLRC1	6	18122171	Lafora disease;not provided
NM_000451.3(SHOX):c.502C>T (p.Arg168Trp)	SHOX	X	640836	Langer mesomelic dysplasia syndrome;Leri Weill dyschondrosteosis
NM_000451.3(SHOX):c.502C>T (p.Arg168Trp)	SHOX	Y	640836	Langer mesomelic dysplasia syndrome;Leri Weill dyschondrosteosis
NM_000451.3(SHOX):c.517C>T (p.Arg173Cys)	SHOX	X	640851	Langer mesomelic dysplasia syndrome;Leri Weill dyschondrosteosis;Short stature, idiopathic, X-linked
NM_000451.3(SHOX):c.517C>T (p.Arg173Cys)	SHOX	Y	640851	Langer mesomelic dysplasia syndrome;Leri Weill dyschondrosteosis;Short stature, idiopathic, X-linked
NM_000163.4(GHR):c.181C>T (p.Arg61Ter)	GHR	5	42688934	Laron-type isolated somatotropin defect
NM_000163.4(GHR):c.703C>T (p.Arg235Ter)	GHR	5	42711291	Laron-type isolated somatotropin defect
NM_000163.4(GHR):c.102G>A (p.Trp34Ter)	GHR	5	42629069	Laron-type isolated somatotropin defect
NM_001457.3(FLNB):c.1081G>A (p.Gly361Ser)	FLNB	3	58097911	Larsen syndrome, dominant type
NM_001457.3(FLNB):c.1088G>A (p.Gly363Glu)	FLNB	3	58097918	Larsen syndrome, dominant type
NM_001457.3(FLNB):c.4808C>T (p.Pro1603Leu)	FLNB	3	58136115	Larsen syndrome, dominant type
NM_001457.3(FLNB):c.5500G>A (p.Gly1834Arg)	FLNB	3	58145995	Larsen syndrome, dominant type
NM_001457.3(FLNB):c.4756G>A (p.Gly1586Arg)	FLNB	3	58136063	Larsen syndrome;Larsen syndrome, dominant type
NM_001457.3(FLNB):c.679G>A (p.Glu227Lys)	FLNB	3	58081668	Larsen syndrome;Larsen syndrome, dominant type
NM_001457.3(FLNB):c.5071G>A (p.Gly1691Ser)	FLNB	3	58138491	Larsen syndrome;Larsen syndrome, dominant type
NM_006918.4(SC5D):c.86G>A (p.Arg29Gln)	SC5D	11	121303461	Lathosterolosis
NM_006918.4(SC5D):c.632G>A (p.Gly211Asp)	SC5D	11	121307244	Lathosterolosis
NM_000358.2(TGFBI):c.370C>T (p.Arg124Cys)	TGFBI	5	136046406	Lattice corneal dystrophy Type I
NM_002353.2(TACSTD2):c.352C>T (p.Gln118Ter)	TACSTD2	1	58576805	Lattice corneal dystrophy Type III
NM_002353.2(TACSTD2):c.619C>T (p.Gln207Ter)	TACSTD2	1	58576538	Lattice corneal dystrophy Type III
NM_025114.3(CEP290):c.1711+1G>A	CEP290	12	88118482	Leber congenital amaurosis 10
NM_000883.3(IMPDH1):c.568C>T (p.Arg190Trp)	IMPDH1	7	128400828	Leber congenital amaurosis 11
NM_183059.2(RD3):c.296+1G>A	RD3	1	211481119	Leber congenital amaurosis 12
NM_183059.2(RD3):c.112C>T (p.Arg38Ter)	RD3	1	211481304	Leber congenital amaurosis 12
NM_152443.2(RDH12):c.565C>T (p.Gln189Ter)	RDH12	14	67727097	Leber congenital amaurosis 13
NM_152443.2(RDH12):c.146C>T (p.Thr49Met)	RDH12	14	67724550	Leber congenital amaurosis 13
NM_152443.2(RDH12):c.184C>T (p.Arg62Ter)	RDH12	14	67724588	Leber congenital amaurosis 13
NM_152443.2(RDH12):c.658+1G>A	RDH12	14	67727191	Leber congenital amaurosis 13
NM_152443.2(RDH12):c.464C>T (p.Thr155Ile)	RDH12	14	67726996	Leber congenital amaurosis 13
NM_003322.5(TULP1):c.1198C>T (p.Arg400Trp)	TULP1	6	35503763	Leber congenital amaurosis 15
NM_003322.5(TULP1):c.1495+1G>A	TULP1	6	35499980	Leber congenital amaurosis 15;Retinitis pigmentosa 14;not provided
NM_002242.4(KCNJ13):c.496C>T (p.Arg166Ter)	KCNJ13	2	232768778	Leber congenital amaurosis 16
NM_002242.4(KCNJ13):c.158G>A (p.Trp53Ter)	KCNJ13	2	232771205	Leber congenital amaurosis 16
NM_001001557.3(GDF6):c.595G>A (p.Ala199Thr)	GDF6	8	96145336	Leber congenital amaurosis 17;Microphthalmia, isolated, with coloboma 6
NM_000329.2(RPE65):c.700C>T (p.Arg234Ter)	RPE65	1	68439586	Leber congenital amaurosis 2;not provided
NM_000329.2(RPE65):c.11+5G>A	RPE65	1	68449890	Leber congenital amaurosis 2;not provided
NM_000329.2(RPE65):c.1543C>T (p.Arg515Trp)	RPE65	1	68429835	Leber congenital amaurosis 2;Retinitis pigmentosa;Retinitis pigmentosa 20;not provided
NM_018418.4(SPATA7):c.1183C>T (p.Arg395Ter)	SPATA7	14	88437565	Leber congenital amaurosis 3
NM_018418.4(SPATA7):c.253C>T (p.Arg85Ter)	SPATA7	14	88416725	Leber congenital amaurosis 3
NM_018418.4(SPATA7):c.322C>T (p.Arg108Ter)	SPATA7	14	88416794	Leber congenital amaurosis 3;SPATA7-Related Disorders
NM_014336.4(AIPL1):c.784G>A (p.Gly262Ser)	AIPL1	17	6426615	Leber congenital amaurosis 4;not provided
NM_020366.3(RPGRIP1):c.1946>A (p.Trp665Ter)	RPGRIP1	14	21294785	Leber congenital amaurosis 6
NM_020366.3(RPGRIP1):c.832C>T (p.Arg278Ter)	RPGRIP1	14	21307762	Leber congenital amaurosis 6

NM_020366.3(RPGRIP1):c.2356C>T (p.Gln786Ter)	RPGRIP1	14	21325372	Leber congenital amaurosis 6
NM_000554.5(CRX):c.268C>T (p.Arg90Trp)	CRX	19	47839335	Leber congenital amaurosis 7;not provided
NM_201253.2(CRB1):c.998G>A (p.Gly333Asp)	CRB1	1	197356840	Leber congenital amaurosis 8
NM_201253.2(CRB1):c.1576C>T (p.Arg526Ter)	CRB1	1	197421404	Leber congenital amaurosis 8;Retinitis pigmentosa 12
NM_201253.2(CRB1):c.3307G>A (p.Gly1103Arg)	CRB1	1	197435170	Leber congenital amaurosis 8;Retinitis pigmentosa 12;not provided
NM_201253.2(CRB1):c.2843G>A (p.Cys948Tyr)	CRB1	1	197434706	Leber congenital amaurosis 8;Retinitis pigmentosa 12;not provided
NM_022787.3(NMNAT1):c.619C>T (p.Arg207Trp)	NMNAT1	1	9982480	Leber congenital amaurosis 9
NM_022787.3(NMNAT1):c.25G>A (p.Val9Met)	NMNAT1	1	9972098	Leber congenital amaurosis 9
NM_022787.3(NMNAT1):c.769G>A (p.Glu257Lys)	NMNAT1	1	9982630	Leber congenital amaurosis 9;not provided
NM_000466.2(PEX1):c.2528G>A (p.Gly843Asp)	PEX1	7	92501562	Leber congenital amaurosis;Peroxisome biogenesis disorder 1B;Peroxisome biogenesis disorders, Zellweger syndrome spectrum;Zellweger syndrome;not provided
m.11696G>A	MT-ND4	MT	11696	Leber hereditary optic neuropathy with dystonia;Leber's optic atrophy
m.14459G>A	MT-ND6	MT	14459	Leber hereditary optic neuropathy with dystonia;Leber's optic atrophy;Leigh syndrome;Leigh syndrome due to mitochondrial complex I deficiency
m.10197G>A	MT-ND3	MT	10197	Leber hereditary optic neuropathy with dystonia;Leigh syndrome;Leigh syndrome due to mitochondrial complex I deficiency
m.9438G>A	MT-CO3	MT	9438	Leber's optic atrophy
m.9804G>A	MT-CO3	MT	9804	Leber's optic atrophy
m.15812G>A	MT-CYB	MT	15812	Leber's optic atrophy
m.13730G>A	MT-ND5	MT	13730	Leber's optic atrophy
m.12848C>T	MT-ND5	MT	12848	Leber's optic atrophy
m.5244G>A	MT-ND2	MT	5244	Leber's optic atrophy
m.3733G>A	MT-ND1	MT	3733	Leber's optic atrophy
m.14568C>T	MT-ND6	MT	14568	Leber's optic atrophy
m.14279G>A	MT-ND6	MT	14279	Leber's optic atrophy
m.14831G>A	MT-CYB	MT	14831	Leber's optic atrophy
m.3635G>A	MT-ND1	MT	3635	Leber's optic atrophy
m.4025C>T	MT-ND1	MT	4025	Leber's optic atrophy
m.3376G>A	MT-ND1	MT	3376	Leber's optic atrophy
NC_012920.1:m.3460G>A	MT-ND1	MT	3460	Leber's optic atrophy;Leigh syndrome;Mitochondrial complex I deficiency
NC_012920.1:m.11778G>A	MT-ND4	MT	11778	Leber's optic atrophy;not provided
NM_001390.4(DTNA):c.362C>T (p.Pro121Leu)	DTNA	18	34794250	Left ventricular noncompaction 1
NM_000257.3(MYH7):c.5296G>A (p.Ala1766Thr)	MYH7	14	23415258	Left ventricular noncompaction 5
NM_000364.3(TNNT2):c.829G>A (p.Asp277Asn)	TNNT2	1	201359636	Left ventricular noncompaction 6
NM_000364.3(TNNT2):c.316G>A (p.Glu106Lys)	TNNT2	1	201365286	Left ventricular noncompaction 6
NM_001001430.2(TNNT2):c.517C>T (p.Arg173Trp)	TNNT2	1	201363349	Left ventricular noncompaction 6;Primary dilated cardiomyopathy;not provided
NM_003240.4(LEFTY2):c.940C>T (p.Arg314Ter)	LEFTY2	1	225937602	Left-right axis malformations
NM_003240.4(LEFTY2):c.1025G>A (p.Ser342Asn)	LEFTY2	1	225937517	Left-right axis malformations
NM_003268.5(TLR5):c.1174C>T (p.Arg392Ter)	TLR5	1	223111858	Legionellosis;Melioidosis, resistance to;Systemic lupus erythematosus, resistance to, 1
NM_152594.2(SPRED1):c.637C>T (p.Gln213Ter)	SPRED1	15	38349476	Legius syndrome
NM_152594.2(SPRED1):c.349C>T (p.Arg117Ter)	SPRED1	15	38322382	Legius syndrome
NM_152594.2(SPRED1):c.70C>T (p.Arg24Ter)	SPRED1	15	38299410	Legius syndrome
NM_152594.2(SPRED1):c.643C>T (p.Gln215Ter)	SPRED1	15	38349482	Legius syndrome
NM_152594.2(SPRED1):c.190C>T (p.Arg64Ter)	SPRED1	15	38299530	Legius syndrome
NM_152594.2(SPRED1):c.973C>T (p.Arg325Ter)	SPRED1	15	38351302	Legius syndrome

NM_152594.2(SPRED1):c.421C>T (p.Gln141Ter)	SPRED1	15	38324807	Legius syndrome
NC_012920.1:m.3481G>A	MT-ND1	MT	3481	Leigh syndrome
NC_012920.1:m.3890G>A	MT-ND1	MT	3890	Leigh syndrome
NC_012920.1:m.10254G>A	MT-ND3	MT	10254	Leigh syndrome
NM_003172.3(SURF1):c.586C>T (p.Gln196Ter)	SURF1	9	133352696	Leigh syndrome
NM_004551.2(NDUFS3):c.434C>T (p.Thr145Ile)	NDUFS3	11	47582140	Leigh syndrome due to mitochondrial complex I deficiency
NM_002495.3(NDUFS4):c.316C>T (p.Arg106Ter)	NDUFS4	5	53646371	Leigh syndrome due to mitochondrial complex I deficiency
NM_024407.4(NDUFS7):c.434G>A (p.Arg145His)	NDUFS7	19	1391144	Leigh syndrome due to mitochondrial complex I deficiency
NM_018838.4(NDUFA12):c.178C>T (p.Arg60Ter)	NDUFA12	12	94994249	Leigh syndrome due to mitochondrial complex I deficiency
NM_004551.2(NDUFS3):c.595C>T (p.Arg199Trp)	NDUFS3	11	47582436	Leigh syndrome due to mitochondrial complex I deficiency;Mitochondrial complex I deficiency
NM_024407.4(NDUFS7):c.364G>A (p.Val122Met)	NDUFS7	19	1391006	Leigh syndrome due to mitochondrial complex I deficiency;not provided
NM_004328.4(BCS1L):c.296C>T (p.Pro99Leu)	BCS1L	2	218661283	Leigh syndrome due to mitochondrial complex III deficiency
NM_133259.3(LRPPRC):c.1061C>T (p.Ala354Val)	LRPPRC	2	43974244	Leigh syndrome, French Canadian type
NM_004092.3(ECHS1):c.5C>T (p.Ala2Val)	ECHS1	10	133373329	Leigh syndrome;Mitochondrial short-chain enoyl-coa hydratase 1 deficiency;not provided
m.1624C>T	MT-TV	MT	1624	Leigh syndrome;Neonatal death
NM_001735.2(C5):c.55C>T (p.Gln19Ter)	C5	9	121050192	Leiner disease
NM_001735.2(C5):c.4426C>T (p.Arg1476Ter)	C5	9	120962749	Leiner disease
NM_014754.2(PTDSS1):c.805C>T (p.Pro269Ser)	PTDSS1	8	96304092	Lenz-Majewski hyperostosis syndrome
NM_002834.4(PTPN11):c.1381G>A (p.Ala461Thr)	PTPN11	12	112488444	LEOPARD syndrome 1;Noonan syndrome with multiple lentigines;not provided
NM_002834.4(PTPN11):c.1492C>T (p.Arg498Trp)	PTPN11	12	112489068	LEOPARD syndrome 1;Rasopathy;not provided
NM_000208.3(INSR):c.1177G>A (p.Gly393Arg)	INSR	19	7172381	Leprechaunism syndrome
NM_000208.3(INSR):c.2095C>T (p.Gln699Ter)	INSR	19	7152862	Leprechaunism syndrome
NM_000208.3(INSR):c.2770C>T (p.Arg924Ter)	INSR	19	7132230	Leprechaunism syndrome
NM_000208.3(INSR):c.172G>A (p.Gly58Arg)	INSR	19	7267825	Leprechaunism syndrome
NM_000208.3(INSR):c.1195C>T (p.Arg399Ter)	INSR	19	7172363	Leprechaunism syndrome
NM_003264.4(TLR2):c.2029C>T (p.Arg677Trp)	TLR2	4	153704936	Leprosy 3
NM_000230.2(LEP):c.313C>T (p.Arg105Trp)	LEP	7	128254572	Leptin deficiency or dysfunction
NM_000451.3(SHOX):c.440G>A (p.Arg147His)	SHOX	X	634780	Leri Weill dyschondrosteosis;Short stature, idiopathic, X-linked
NM_000451.3(SHOX):c.440G>A (p.Arg147His)	SHOX	Y	634780	Leri Weill dyschondrosteosis;Short stature, idiopathic, X-linked
NM_000194.2(HPRT1):c.580G>A (p.Asp194Asn)	HPRT1	X	134498655	Lesch-Nyhan syndrome
NM_000194.2(HPRT1):c.209G>A (p.Gly70Glu)	HPRT1	X	134475255	Lesch-Nyhan syndrome
NM_000194.2(HPRT1):c.325C>T (p.Gln109Ter)	HPRT1	X	134486471	Lesch-Nyhan syndrome
NM_000194.2(HPRT1):c.134G>A (p.Arg45Lys)	HPRT1	X	134473465	Lesch-Nyhan syndrome
NM_000194.2(HPRT1):c.527C>T (p.Pro176Leu)	HPRT1	X	134498431	Lesch-Nyhan syndrome
NM_000194.2(HPRT1):c.508C>T (p.Arg170Ter)	HPRT1	X	134498412	Lesch-Nyhan syndrome
NM_000194.2(HPRT1):c.419G>A (p.Gly140Asp)	HPRT1	X	134493524	Lesch-Nyhan syndrome
NM_000194.2(HPRT1):c.143G>A (p.Arg48His)	HPRT1	X	134475189	Lesch-nyhan syndrome, neurologic variant;Partial hypoxanthine-guanine phosphoribosyltransferase deficiency
NM_000194.2(HPRT1):c.151C>T (p.Arg51Ter)	HPRT1	X	134475197	Lesch-Nyhan syndrome;not provided
NM_000194.2(HPRT1):c.384+1G>A	HPRT1	X	134486531	Lesch-Nyhan syndrome;not provided
NM_001003722.1(GLE1):c.1849G>A (p.Val617Met)	GLE1	9	128538058	Lethal arthrogryposis with anterior horn cell disease
NM_012398.2(PIP5K1C):c.757G>A (p.Asp253Asn)	PIP5K1C	19	3653454	Lethal congenital contracturai syndrome 3
NM_001003722.1(GLE1):c.1706G>A (p.Arg569His)	GLE1	9	128536414	Lethal congenital contracture syndrome 1
NM_033116.5(NEK9):c.1489C>T (p.Arg497Ter)	NEK9	14	75106541	Lethal congenital contracture syndrome 10
NM_181789.2(GLDN):c.1240C>T (p.Arg414Ter)	GLDN	15	51404338	LETHAL CONGENITAL CONTRACTURE SYNDROME 11

NM_181789.2(GLDN):c.541+1G>A	GLDN	15	51383893	LETHAL CONGENITAL CONTRACTURE SYNDROME 11
NM_181789.2(GLDN):c.1435C>T (p.Arg479Ter)	GLDN	15	51404533	LETHAL CONGENITAL CONTRACTURE SYNDROME 11
NM_002465.3(MYBPC1):c.952C>T (p.Arg318Ter)	MYBPC1	12	101644783	Lethal congenital contracture syndrome 4
NM_001137601.2(ZBTB42):c.1190G>A (p.Arg397His)	ZBTB42	14	104802387	Lethal congenital contracture syndrome 6
NM_015270.4(ADCY6):c.3346C>T (p.Arg1116Cys)	ADCY6	12	48768972	Lethal congenital contracture syndrome 8
NM_005529.6(HSPG2):c.10894C>T (p.Arg3632Ter)	HSPG2	1	21833551	Lethal Kniest-like syndrome;Schwartz Jampel syndrome type 1
NM_000751.2(CHRND):c.234G>A (p.Trp78Ter)	CHRND	2	232527436	Lethal multiple pterygium syndrome
NM_000751.2(CHRND):c.1390C>T (p.Arg464Ter)	CHRND	2	232535148	Lethal multiple pterygium syndrome
NM_005199.4(CHRNG):c.715C>T (p.Arg239Cys)	CHRNG	2	232542992	Lethal multiple pterygium syndrome;Multiple pterygium syndrome Escobar type
NM_001287174.1(ABCC8):c.4058G>A (p.Arg1353His)	ABCC8	11	17396980	Leucine-induced hypoglycemia
NM_001280547.1(PAX5):c.547G>A (p.Gly183Ser)	PAX5	9	37002705	Leukemia, acute lymphoblastic, susceptibility to, 3
NM_000211.4(ITGB2):c.1777C>T (p.Arg593Cys)	ITGB2	21	44889376	Leukocyte adhesion deficiency
NM_000211.4(ITGB2):c.505G>A (p.Gly169Arg)	ITGB2	21	44901728	Leukocyte adhesion deficiency
NM_000211.4(ITGB2):c.533C>T (p.Pro178Leu)	ITGB2	21	44901700	Leukocyte adhesion deficiency
NM_000211.4(ITGB2):c.382G>A (p.Asp128Asn)	ITGB2	21	44903482	Leukocyte adhesion deficiency
NM_000211.4(ITGB2):c.850G>A (p.Gly284Ser)	ITGB2	21	44900367	Leukocyte adhesion deficiency
NM_000211.4(ITGB2):c.897+1G>A	ITGB2	21	44900319	Leukocyte adhesion deficiency type 1
NM_000211.4(ITGB2):c.715G>A (p.Ala239Thr)	ITGB2	21	44901518	Leukocyte adhesion deficiency type 1;not provided
NM_178443.2(FERMT3):c.48G>A (p.Trp16Ter)	FERMT3	11	64207412	Leukocyte adhesion deficiency, type III
NM_178443.2(FERMT3):c.1729C>T (p.Arg577Ter)	FERMT3	11	64223094	Leukocyte adhesion deficiency, type III
NM_178443.2(FERMT3):c.687G>A (p.Trp229Ter)	FERMT3	11	64211648	Leukocyte adhesion deficiency, type III
NM_178443.2(FERMT3):c.1537C>T (p.Arg513Ter)	FERMT3	11	64220649	Leukocyte adhesion deficiency, type III
NM_000211.4(ITGB2):c.817G>A (p.Gly273Arg)	ITGB2	21	44900400	Leukocyte adhesion deficiency;not provided
NM_001142415.1(AIMP1):c.115C>T (p.Gln39Ter)	AIMP1	4	106327456	Leukodystrophy, hypomyelinating 3
NM_001142415.1(AIMP1):c.334C>T (p.Gln112Ter)	AIMP1	4	106328186	Leukodystrophy, hypomyelinating 3
NM_013328.3(PYCR2):c.355C>T (p.Arg119Cys)	PYCR2	1	225922043	Leukodystrophy, hypomyelinating, 10
NM_013328.3(PYCR2):c.751C>T (p.Arg251Cys)	PYCR2	1	225921254	Leukodystrophy, hypomyelinating, 10
NM_013328.3(PYCR2):c.796C>T (p.Arg266Ter)	PYCR2	1	225921209	Leukodystrophy, hypomyelinating, 10
NM_013328.3(PYCR2):c.595C>T (p.Arg199Trp)	PYCR2	1	225921590	Leukodystrophy, hypomyelinating, 10
NM_203290.3(POLR1C):c.77C>T (p.Thr26Ile)	POLR1C	6	43517313	Leukodystrophy, hypomyelinating, 11
NM_203290.3(POLR1C):c.326G>A (p.Arg109His)	POLR1C	6	43519782	Leukodystrophy, hypomyelinating, 11
NM_203290.3(POLR1C):c.88C>T (p.Pro30Ser)	POLR1C	6	43517324	Leukodystrophy, hypomyelinating, 11
NM_020435.3(GJC2):c.268C>T (p.Pro90Ser)	GJC2	1	228158026	Leukodystrophy, hypomyelinating, 2
NM_020435.3(GJC2):c.718C>T (p.Arg240Ter)	GJC2	1	228158476	Leukodystrophy, hypomyelinating, 2
NM_020435.3(GJC2):c.787G>A (p.Glu263Lys)	GJC2	1	228158545	Leukodystrophy, hypomyelinating, 2
NM_006087.3(TUBB4A):c.1228G>A (p.Glu410Lys)	TUBB4A	19	6495271	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.568C>T (p.His190Tyr)	TUBB4A	19	6495931	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.1172G>A (p.R391H)	TUBB4A	19	6495327	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.1164G>A (p.M388I)	TUBB4A	19	6495335	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.1061G>A (p.C354Y)	TUBB4A	19	6495438	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.1054G>A (p.A352T)	TUBB4A	19	6495445	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.731G>A (p.G244D)	TUBB4A	19	6495768	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.730G>A (p.Gly244Ser)	TUBB4A	19	6495769	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.533C>T (p.T178M)	TUBB4A	19	6495966	Leukodystrophy, hypomyelinating, 6
NM_006087.3(TUBB4A):c.4C>T (p.R2W)	TUBB4A	19	6502209	Leukodystrophy, hypomyelinating, 6

NM_006087.3(TUBB4A):c.745G>A (p.Asp249Asn)	TUBB4A	19	6495754	Leukodystrophy, hypomyelinating, 6;not provided
NM_006087.3(TUBB4A):c.5G>A (p.Arg2Gln)	TUBB4A	19	6502208	Leukodystrophy, hypomyelinating, 6;not provided
NM_006087.3(TUBB4A):c.763G>A (p.Val255Ile)	TUBB4A	19	6495736	Leukodystrophy, hypomyelinating, 6;not provided
NM_006087.3(TUBB4A):c.785G>A (p.Arg262His)	TUBB4A	19	6495714	Leukodystrophy, hypomyelinating, 6;not provided
NM_006087.3(TUBB4A):c.941C>T (p.A314V)	TUBB4A	19	6495558	Leukodystrophy, hypomyelinating, 6;not provided
NM_002887.3(RARS):c.1535G>A (p.Arg512Gln)	RARS	5	168516860	Leukodystrophy, hypomyelinating, 9
NM_002887.3(RARS):c.1367C>T (p.Ser456Leu)	RARS	5	168510601	Leukodystrophy, hypomyelinating, 9
NM_004366.5(CLCN2):c.1709G>A (p.Trp570Ter)	CLCN2	3	184354113	Leukoencephalopathy with ataxia
NM_004366.5(CLCN2):c.1499C>T (p.Ala500Val)	CLCN2	3	184354556	Leukoencephalopathy with ataxia
NM_004366.5(CLCN2):c.2386C>T (p.Gln796Ter)	CLCN2	3	184352042	Leukoencephalopathy with ataxia
NM_004366.5(CLCN2):c.1795G>A (p.Asp599Asn)	CLCN2	3	184353722	Leukoencephalopathy with ataxia
NM_004366.5(CLCN2):c.1507G>A (p.Gly503Arg)	CLCN2	3	184354548	Leukoencephalopathy with ataxia
NM_004366.5(CLCN2):c.1412G>A (p.Arg471His)	CLCN2	3	184354643	Leukoencephalopathy with ataxia
NM_004366.5(CLCN2):c.1397G>A (p.Gly466Glu)	CLCN2	3	184354658	Leukoencephalopathy with ataxia
NM_004366.5(CLCN2):c.925C>T (p.Arg309Ter)	CLCN2	3	184357240	Leukoencephalopathy with ataxia
NM_018122.4(DARS2):c.787C>T (p.Arg263Ter)	DARS2	1	173838206	Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation
NM_018122.4(DARS2):c.788G>A (p.Arg263Gln)	DARS2	1	173838207	Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation
NM_018122.4(DARS2):c.536G>A (p.Arg179His)	DARS2	1	173833419	Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation
NM_018122.4(DARS2):c.1837C>T (p.Leu613Phe)	DARS2	1	173857604	Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation
NM_018122.4(DARS2):c.1825C>T (p.Arg609Trp)	DARS2	1	173857592	Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation
NM_001034116.1(EIF2B4):c.1070G>A (p.Arg357Gln)	EIF2B4	2	27366880	Leukoencephalopathy with vanishing white matter
NM_001034116.1(EIF2B4):c.1120C>T (p.Arg374Cys)	EIF2B4	2	27366830	Leukoencephalopathy with vanishing white matter
NM_001034116.1(EIF2B4):c.1191+1G>A	EIF2B4	2	27366758	Leukoencephalopathy with vanishing white matter
NM_001034116.1(EIF2B4):c.683C>T (p.Ala228Val)	EIF2B4	2	27368047	Leukoencephalopathy with vanishing white matter
NM_020365.4(EIF2B3):c.674G>A (p.Arg225Gln)	EIF2B3	1	44881722	Leukoencephalopathy with vanishing white matter
NM_020365.4(EIF2B3):c.260C>T (p.Ala87Val)	EIF2B3	1	44978349	Leukoencephalopathy with vanishing white matter
NM_003907.2(EIF2B5):c.584G>A (p.Arg195His)	EIF2B5	3	184137975	Leukoencephalopathy with vanishing white matter
NM_003907.2(EIF2B5):c.944G>A (p.Arg315His)	EIF2B5	3	184140518	Leukoencephalopathy with vanishing white matter
NM_003907.2(EIF2B5):c.241G>A (p.Glu81Lys)	EIF2B5	3	184136657	Leukoencephalopathy with vanishing white matter
NM_003907.2(EIF2B5):c.545C>T (p.Thr182Met)	EIF2B5	3	184137936	Leukoencephalopathy with vanishing white matter;not provided
NM_003907.2(EIF2B5):c.338G>A (p.Arg113His)	EIF2B5	3	184137637	Leukoencephalopathy with vanishing white matter;Ovarioleukodystrophy;not provided
NR_033294.1(SNORD118):n.*5C>G	SNORD118	17	8173448	Leukoencephalopathy, brain calcifications, and cysts
NR_033294.1(SNORD118):n.57G>A	SNORD118	17	8173532	Leukoencephalopathy, brain calcifications, and cysts
NR_033294.1(SNORD118):n.145C>T	SNORD118	17	8173444	Leukoencephalopathy, brain calcifications, and cysts
NM_020745.3(AARS2):c.1561C>T (p.Arg521Ter)	AARS2	6	44305072	Leukoencephalopathy, progressive, with ovarian failure
NM_020745.3(AARS2):c.2893G>A (p.Gly965Arg)	AARS2	6	44300612	Leukoencephalopathy, progressive, with ovarian failure
NM_020745.3(AARS2):c.1213G>A (p.Glu405Lys)	AARS2	6	44306367	Leukoencephalopathy, progressive, with ovarian failure
NM_006225.3(PLCD1):c.1246C>T (p.Arg416Ter)	PLCD1	3	38009945	Leukonychia totalis
NM_006225.3(PLCD1):c.1657G>A (p.Ala553Thr)	PLCD1	3	38009108	Leukonychia totalis
NM_000141.4(FGFR2):c.1942G>A (p.Ala648Thr)	FGFR2	10	121488035	Levy-Hollister syndrome
NM_000142.4(FGFR3):c.1537G>A (p.Asp513Asn)	FGFR3	4	1805561	Levy-Hollister syndrome
NM_000141.4(FGFR2):c.1882G>A (p.Ala628Thr)	FGFR2	10	121488095	Levy-Hollister syndrome;not provided
NM_001001502.2(SNCB):c.208G>A (p.Val70Met)	SNCB	5	176626472	Lewy body dementia
NM_000345.3(SNCA):c.136G>A (p.Glu46Lys)	SNCA	4	89828170	Lewy body dementia
NM_000233.3(LHCGR):c.1660C>T (p.Arg554Ter)	LHCGR	2	48688137	Leydig cell agenesis;Luteinizing hormone resistance, female

NM_000233.3(LHCGR):c.1060G>A (p.Glu354Lys)	LHCGR	2	48688737	Leydig cell agenesis;Luteinizing hormone resistance, female
NM_003047.4(SLC9A1):c.913G>A (p.Gly305Arg)	SLC9A1	1	27109678	Lichtenstein-knorr syndrome
NM_000546.5(TP53):c.672+1G>A	TP53	17	7674858	Li-Fraumeni syndrome
NM_000546.5(TP53):c.574C>T (p.Gln192Ter)	TP53	17	7674957	Li-Fraumeni syndrome
NM_000546.5(TP53):c.772G>A (p.Glu258Lys)	TP53	17	7674191	Li-Fraumeni syndrome 1;not provided
NM_002312.3(LIG4):c.1738C>T (p.Arg580Ter)	LIG4	13	108209531	Lig4 syndrome
NM_002312.3(LIG4):c.1406G>A (p.Gly469Glu)	LIG4	13	108209863	Lig4 syndrome
NM_002312.3(LIG4):c.833G>A (p.Arg278His)	LIG4	13	108210436	Lig4 syndrome
NM_002312.3(LIG4):c.2440C>T (p.Arg814Ter)	LIG4	13	108208829	Lig4 syndrome;not provided
NM_000232.4(SGCB):c.31C>T (p.Gln11Ter)	SGCB	4	52038229	Limb-Girdle Muscular Dystrophy, Recessive;Limb-girdle muscular dystrophy, type 2E
NM_006790.2(MYOT):c.17G>A (p.Arg6His)	MYOT	5	137870668	Limb-girdle muscular dystrophy, type 1A
NM_006790.2(MYOT):c.164C>T (p.Ser55Phe)	MYOT	5	137870815	Limb-girdle muscular dystrophy, type 1A;Myotilinopathy;Progressive distal muscle weakness;Progressive proximal muscle weakness
NM_006790.2(MYOT):c.170C>T (p.Thr57Ile)	MYOT	5	137870821	Limb-girdle muscular dystrophy, type 1A;not provided
NM_170707.3(LMNA):c.1488+1G>A	LMNA	1	156137029	Limb-girdle muscular dystrophy, type 1B;Mandibuloacral dysostosis;not provided
NM_170707.3(LMNA):c.1477C>T (p.Gln493Ter)	LMNA	1	156137017	Limb-girdle muscular dystrophy, type 1B;not provided
NM_170707.3(LMNA):c.1609-1G>A	LMNA	1	156137653	Limb-girdle muscular dystrophy, type 1B;Primary dilated cardiomyopathy;not provided
NM_033337.2(CAV3):c.314C>T (p.Pro105Leu)	CAV3	3	8745725	Limb-girdle muscular dystrophy, type 1C;Rippling muscle disease 2;not provided
NM_058246.3(DNAJB6):c.346+5G>A	DNAJB6	7	157367488	Limb-girdle muscular dystrophy, type 1E
NM_031372.3(HNRNPDL):c.1132G>A (p.Asp378Asn)	HNRNPDL	4	82426523	Limb-girdle muscular dystrophy, type 1G
NM_000070.2(CAPN3):c.2306G>A (p.Arg769Gln)	CAPN3	15	42410926	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1715G>A (p.Arg572Gln)	CAPN3	15	42402972	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.328C>T (p.Arg110Ter)	CAPN3	15	42384501	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.257C>T (p.Ser86Phe)	CAPN3	15	42360062	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.946-1G>A	CAPN3	15	42392638	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.2243G>A (p.Arg748Gln)	CAPN3	15	42410646	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.145C>T (p.Arg49Cys)	CAPN3	15	42359950	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1319G>A (p.Arg440Gln)	CAPN3	15	42399617	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.133G>A (p.Ala45Thr)	CAPN3	15	42359938	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.499-1G>A	CAPN3	15	42387752	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1343G>A (p.Arg448His)	CAPN3	15	42399641	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1465C>T (p.Arg489Trp)	CAPN3	15	42401751	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1714C>T (p.Arg572Trp)	CAPN3	15	42402971	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.439C>T (p.Arg147Ter)	CAPN3	15	42386226	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1063C>T (p.Arg355Trp)	CAPN3	15	42394289	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1250C>T (p.Thr417Met)	CAPN3	15	42399548	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.245C>T (p.Pro82Leu)	CAPN3	15	42360050	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.2242C>T (p.Arg748Ter)	CAPN3	15	42410645	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1318C>T (p.Arg440Trp)	CAPN3	15	42399616	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1333G>A (p.Gly445Arg)	CAPN3	15	42399631	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1957C>T (p.Gln653Ter)	CAPN3	15	42409345	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.1801-1G>A	CAPN3	15	42408210	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.2263+1G>A	CAPN3	15	42410667	Limb-girdle muscular dystrophy, type 2A
NM_000070.2(CAPN3):c.956C>T (p.Pro319Leu)	CAPN3	15	42392649	Limb-girdle muscular dystrophy, type 2A;not provided
NM_000070.2(CAPN3):c.1468C>T (p.Arg490Trp)	CAPN3	15	42401754	Limb-girdle muscular dystrophy, type 2A;not provided

NM_000070.2(CAPN3):c.802-9G>A	CAPN3	15	42389944	Limb-girdle muscular dystrophy, type 2A;not provided
NM_000070.2(CAPN3):c.1342C>T (p.Arg448Cys)	CAPN3	15	42399640	Limb-girdle muscular dystrophy, type 2A;not provided
NM_000070.2(CAPN3):c.1303G>A (p.Glu435Lys)	CAPN3	15	42399601	Limb-girdle muscular dystrophy, type 2A;not provided
NM_000070.2(CAPN3):c.1993-1G>A	CAPN3	15	42409786	Limb-girdle muscular dystrophy, type 2A;not provided
NM_003494.3(DYSF):c.5057+5G>A	DYSF	2	71664443	Limb-girdle muscular dystrophy, type 2B
NM_001130978.1(DYSF):c.895G>A (p.Gly299Arg)	DYSF	2	71517028	Limb-girdle muscular dystrophy, type 2B
NM_003494.3(DYSF):c.3113G>A (p.Arg1038Gln)	DYSF	2	71570680	Limb-girdle muscular dystrophy, type 2B
NM_001130987.1(DYSF):c.5174+1G>A	DYSF	2	71664439	Limb-girdle muscular dystrophy, type 2B
NM_001130987.1(DYSF):c.159G>A (p.Trp53Ter)	DYSF	2	71481890	Limb-girdle muscular dystrophy, type 2B
NM_001130987.1(DYSF):c.2929C>T (p.Arg977Trp)	DYSF	2	71569884	Limb-girdle muscular dystrophy, type 2B
NM_001130987.1(DYSF):c.4282C>T (p.Gln1428Ter)	DYSF	2	71612701	Limb-girdle muscular dystrophy, type 2B
NM_001130987.1(DYSF):c.1577-1G>A	DYSF	2	71551040	Limb-girdle muscular dystrophy, type 2B
NM_003494.3(DYSF):c.5529G>A (p.Trp1843Ter)	DYSF	2	71669608	Limb-girdle muscular dystrophy, type 2B
NM_001130987.1(DYSF):c.1576+1G>A	DYSF	2	71539240	Limb-girdle muscular dystrophy, type 2B
NM_001130987.1(DYSF):c.4462C>T (p.Gln1488Ter)	DYSF	2	71613408	Limb-girdle muscular dystrophy, type 2B
NM_001130978.1(DYSF):c.1813C>T (p.Gln605Ter)	DYSF	2	71553071	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.5429G>A (p.Arg1810Lys)	DYSF	2	71668842	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.3230G>A (p.Trp1077Ter)	DYSF	2	71574253	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.265C>T (p.Arg89Ter)	DYSF	2	71503242	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.4434G>A (p.Trp1478Ter)	DYSF	2	71643988	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.5077C>T (p.Arg1693Trp)	DYSF	2	71665181	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.3478C>T (p.Gln1160Ter)	DYSF	2	71590246	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_001130987.1(DYSF):c.1372G>A (p.Gly458Arg)	DYSF	2	71528393	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.4090C>T (p.Gln1364Ter)	DYSF	2	71611549	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_001130987.1(DYSF):c.2409+1G>A	DYSF	2	71561945	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.1708C>T (p.Gln570Ter)	DYSF	2	71551676	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1
NM_003494.3(DYSF):c.1956G>A (p.Trp652Ter)	DYSF	2	71553832	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;Myopathy, distal, with anterior tibial onset
NM_001130987.1(DYSF):c.5004-1G>A	DYSF	2	71664267	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;Myopathy, distal, with anterior tibial onset
NM_001130978.1(DYSF):c.5776C>T (p.Arg1926Ter)	DYSF	2	71674242	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;Myopathy, distal, with anterior tibial onset;not provided
NM_003494.3(DYSF):c.331C>T (p.Gln111Ter)	DYSF	2	71503308	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;Myopathy, distal, with anterior tibial onset;not provided
NM_003494.3(DYSF):c.6124C>T (p.Arg2042Cys)	DYSF	2	71682597	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_001130978.1(DYSF):c.3137G>A (p.Arg1046His)	DYSF	2	71570704	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.1053+1G>A	DYSF	2	71520905	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.1398-1G>A	DYSF	2	71539156	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.1481-1G>A	DYSF	2	71549349	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.2311C>T (p.Gln771Ter)	DYSF	2	71561900	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.2643+1G>A	DYSF	2	71568083	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.2869C>T (p.Gln957Ter)	DYSF	2	71569878	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.4253G>A (p.Gly1418Asp)	DYSF	2	71612726	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.4756C>T (p.Arg1586Ter)	DYSF	2	71658995	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.5509G>A (p.Asp1837Asn)	DYSF	2	71669191	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided

NM_003494.3(DYSF):c.5644C>T (p.Gln1882Ter)	DYSF	2	71669723	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.5946+1G>A	DYSF	2	71679236	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.610C>T (p.Arg204Ter)	DYSF	2	71513868	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.937+1G>A	DYSF	2	71520209	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.5266C>T (p.Gln1756Ter)	DYSF	2	71667441	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.3832C>T (p.Gln1278Ter)	DYSF	2	71600831	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.5525+1G>A	DYSF	2	71669208	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.1834C>T (p.Gln612Ter)	DYSF	2	71553092	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.5668-7G>A	DYSF	2	71674190	Limb-girdle muscular dystrophy, type 2B;Miyoshi muscular dystrophy 1;not provided
NM_003494.3(DYSF):c.3112C>T (p.Arg1038Ter)	DYSF	2	71570679	Limb-girdle muscular dystrophy, type 2B;not provided
NM_000023.3(SGCA):c.293G>A (p.Arg98His)	SGCA	17	50167717	Limb-girdle muscular dystrophy, type 2D
NM_000023.3(SGCA):c.850C>T (p.Arg284Cys)	SGCA	17	50170245	Limb-girdle muscular dystrophy, type 2D
NM_000023.3(SGCA):c.574C>T (p.Arg192Ter)	SGCA	17	50168562	Limb-girdle muscular dystrophy, type 2D
NM_000023.3(SGCA):c.403C>T (p.Gln135Ter)	SGCA	17	50168391	Limb-girdle muscular dystrophy, type 2D
NM_000023.3(SGCA):c.409G>A (p.Glu137Lys)	SGCA	17	50168397	Limb-girdle muscular dystrophy, type 2D
NM_000023.3(SGCA):c.747+1G>A	SGCA	17	50169255	Limb-girdle muscular dystrophy, type 2D
NM_000023.3(SGCA):c.229C>T (p.Arg77Cys)	SGCA	17	50167653	Limb-girdle muscular dystrophy, type 2D;not provided
NM_000023.3(SGCA):c.101G>A (p.Arg34His)	SGCA	17	50167431	Limb-girdle muscular dystrophy, type 2D;not provided
NM_000023.3(SGCA):c.739G>A (p.Val247Met)	SGCA	17	50169246	Limb-girdle muscular dystrophy, type 2D;not provided
NM_000337.5(SGCD):c.493C>T (p.Arg165Ter)	SGCD	5	156595042	Limb-girdle muscular dystrophy, type 2F
NM_000337.5(SGCD):c.89G>A (p.Trp30Ter)	SGCD	5	156344574	Limb-girdle muscular dystrophy, type 2F
NM_000337.5(SGCD):c.784G>A (p.Glu262Lys)	SGCD	5	156759301	Limb-girdle muscular dystrophy, type 2F
NM_001256850.1(TTN):c.87394C>T (p.Arg29132Ter)	TTN	2	178549309	Limb-girdle muscular dystrophy, type 2J;not provided
NM_213599.2(ANO5):c.1407+5G>A	ANO5	11	22257759	Limb-girdle muscular dystrophy, type 2L
NM_213599.2(ANO5):c.762+1G>A	ANO5	11	22236277	Limb-girdle muscular dystrophy, type 2L
NM_213599.2(ANO5):c.1213C>T (p.Gln405Ter)	ANO5	11	22255403	Limb-girdle muscular dystrophy, type 2L
NM_213599.2(ANO5):c.1639C>T (p.Arg547Ter)	ANO5	11	22262137	Limb-girdle muscular dystrophy, type 2L
NM_213599.2(ANO5):c.1406G>A (p.Trp469Ter)	ANO5	11	22257753	Limb-girdle muscular dystrophy, type 2L
NM_213599.2(ANO5):c.1210C>T (p.Arg404Ter)	ANO5	11	22255400	Limb-girdle muscular dystrophy, type 2L;Miyoshi muscular dystrophy 3
NM_213599.2(ANO5):c.2272C>T (p.Arg758Cys)	ANO5	11	22274605	Limb-girdle muscular dystrophy, type 2L;Miyoshi muscular dystrophy 3;not provided
NM_213599.2(ANO5):c.41-1G>A	ANO5	11	22203803	Limb-girdle muscular dystrophy, type 2L;Miyoshi muscular dystrophy 3;not provided
NM_213599.2(ANO5):c.172C>T (p.Arg58Trp)	ANO5	11	22218279	Limb-girdle muscular dystrophy, type 2L;Miyoshi muscular dystrophy 3;not provided
NM_213599.2(ANO5):c.1898+1G>A	ANO5	11	22263044	Limb-girdle muscular dystrophy, type 2L;not provided
NM_021942.5(TRAPPC11):c.2938G>A (p.Gly980Arg)	TRAPPC11	4	183701783	Limb-girdle muscular dystrophy, type 2S
NM_199053.2(TRAPPC11):c.142C>T (p.Arg48Ter)	TRAPPC11	4	183664009	Limb-girdle muscular dystrophy, type 2S
NM_021942.5(TRAPPC11):c.1287+5G>A	TRAPPC11	4	183684059	Limb-girdle muscular dystrophy, type 2S;not provided
NM_170707.3(LMNA):c.1608+1G>A	LMNA	1	156137233	Limb-girdle muscular dystrophy;not provided
NM_007171.3(POMT1):c.1241C>T (p.Thr414Met)	POMT1	9	131513331	Limb-girdle muscular dystrophy-dystroglycanopathy, type C1
NM_007171.3(POMT1):c.1864C>T (p.Arg622Ter)	POMT1	9	131521445	Limb-girdle muscular dystrophy-dystroglycanopathy, type C1;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 1
NM_001079802.1(FKTN):c.340G>A (p.Ala114Thr)	FKTN	9	105601319	Limb-girdle muscular dystrophy-dystroglycanopathy, type C4
NM_024301.4(FKRP):c.160C>T (p.Arg54Trp)	FKRP	19	46755610	Limb-girdle muscular dystrophy-dystroglycanopathy, type C5
NM_024301.4(FKRP):c.764G>A (p.Trp255Ter)	FKRP	19	46756214	Limb-girdle muscular dystrophy-dystroglycanopathy, type C5
NM_024301.4(FKRP):c.400C>T (p.Arg134Trp)	FKRP	19	46755850	Limb-girdle muscular dystrophy-dystroglycanopathy, type C5
NM_024301.4(FKRP):c.313C>T (p.Gln105Ter)	FKRP	19	46755763	Limb-girdle muscular dystrophy-dystroglycanopathy, type C5

NM_001165928.3(DAG1):c.220G>A (p.Val74Ile)	DAG1	3	49510754	Limb-girdle muscular dystrophy-dystroglycanopathy, type C9
NM_001165928.3(DAG1):c.575C>T (p.Thr192Met)	DAG1	3	49531086	Limb-girdle muscular dystrophy-dystroglycanopathy, type C9;not provided
NM_005333.4(HCCS):c.589C>T (p.Arg197Ter)	HCCS	X	11120974	Linear skin defects with multiple congenital anomalies 1
NM_005333.4(HCCS):c.649C>T (p.Arg217Cys)	HCCS	X	11121652	Linear skin defects with multiple congenital anomalies 1
NM_005333.4(HCCS):c.475G>A (p.Glu159Lys)	HCCS	X	11118574	Linear skin defects with multiple congenital anomalies 1
NM_001866.2(COX7B):c.55C>T (p.Gln19Ter)	COX7B	X	77902657	Linear skin defects with multiple congenital anomalies 2
NM_022773.2(LMF1):c.1391G>A (p.Trp464Ter)	LMF1	16	869908	Lipase deficiency combined
NM_004425.3(ECM1):c.1036C>T (p.Gln346Ter)	ECM1	1	150511784	Lipid proteinosis
NM_004425.3(ECM1):c.157C>T (p.Arg53Ter)	ECM1	1	150509696	Lipid proteinosis
NM_004425.3(ECM1):c.727C>T (p.Arg243Ter)	ECM1	1	150511475	Lipid proteinosis
NM_004425.3(ECM1):c.826C>T (p.Gln276Ter)	ECM1	1	150511574	Lipid proteinosis
NM_001753.4(CAV1):c.424C>T (p.Gln142Ter)	CAV1	7	116559174	Lipodystrophy, congenital generalized, type 3
NM_000041.3(APOE):c.127C>T (p.Arg43Cys)	APOE	19	44907843	Lipoprotein glomerulopathy
NM_005577.2(LPA):c.109C>T (p.Arg37Ter)	LPA	6	160650438	Lipoprotein(a) deficiency, congenital
NM_015929.3(LIPT1):c.212C>T (p.Ser71Phe)	LIPT1	2	99162169	Lipoyltransferase 1 deficiency
NM_000430.3(PAFAH1B1):c.484G>A (p.Gly162Ser)	PAFAH1B1	17	2670247	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.1002+1G>A	PAFAH1B1	17	2676607	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.3G>A (p.Met1Ile)	PAFAH1B1	17	2638291	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.33-3C>T	PAFAH1B1	17	2665369	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.37C>T (p.Arg13Ter)	PAFAH1B1	17	2665376	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.192+1G>A	PAFAH1B1	17	2666091	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.399+1G>A	PAFAH1B1	17	2667199	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.405G>A (p.Trp135Ter)	PAFAH1B1	17	2670168	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.460C>T (p.Gln154Ter)	PAFAH1B1	17	2670223	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.657G>A (p.Trp219Ter)	PAFAH1B1	17	2672743	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.664C>T (p.Gln222Ter)	PAFAH1B1	17	2672750	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.671G>A (p.Gly224Asp)	PAFAH1B1	17	2672757	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.730C>T (p.Gln244Ter)	PAFAH1B1	17	2674118	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.851G>A (p.Trp284Ter)	PAFAH1B1	17	2674239	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.900+1G>A	PAFAH1B1	17	2674289	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.1002+5G>A	PAFAH1B1	17	2676611	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.1009C>T (p.His337Tyr)	PAFAH1B1	17	2680170	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.1064G>A (p.Ser355Asn)	PAFAH1B1	17	2680225	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.1111C>T (p.Arg371Ter)	PAFAH1B1	17	2680272	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.1135C>T (p.His379Tyr)	PAFAH1B1	17	2680296	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.1165C>T (p.His389Tyr)	PAFAH1B1	17	2681734	Lissencephaly 1
NM_000430.3(PAFAH1B1):c.817C>T (p.Arg273Ter)	PAFAH1B1	17	2674205	Lissencephaly 1;not provided
NM_000430.3(PAFAH1B1):c.265C>T (p.Arg89Ter)	PAFAH1B1	17	2667064	Lissencephaly 1;not provided
NM_000430.3(PAFAH1B1):c.430C>T (p.Arg144Ter)	PAFAH1B1	17	2670193	Lissencephaly 1;not provided
NM_000430.3(PAFAH1B1):c.22C>T (p.Arg8Ter)	PAFAH1B1	17	2638310	Lissencephaly 1;Subcortical band heterotopia
NM_005045.3(RELN):c.5969+1G>A	RELN	7	103553659	Lissencephaly 2
NM_005045.3(RELN):c.7490+1G>A	RELN	7	103523390	Lissencephaly 2 (Norman-Roberts type)
NM_005045.3(RELN):c.5587C>T (p.Gln1863Ter)	RELN	7	103557992	Lissencephaly 2 (Norman-Roberts type)
NM_139058.2(ARX):c.1117C>T (p.Gln373Ter)	ARX	X	25010262	Lissencephaly 2, X-linked
NM_139058.2(ARX):c.1414C>T (p.Arg472Ter)	ARX	X	25007145	Lissencephaly 2, X-linked

NM_139058.2(ARX):c.995G>A (p.Arg332His)	ARX	X	25013000	Lissencephaly 2, X-linked;not provided
NM_006009.3(TUBA1A):c.790C>T (p.Arg264Cys)	TUBA1A	12	49185576	Lissencephaly 3
NM_006009.3(TUBA1A):c.1205G>A (p.Arg402His)	TUBA1A	12	49185161	Lissencephaly 3
NM_006009.3(TUBA1A):c.1256C>T (p.Ser419Leu)	TUBA1A	12	49185110	Lissencephaly 3
NM_006009.3(TUBA1A):c.1204C>T (p.Arg402Cys)	TUBA1A	12	49185162	Lissencephaly 3
NM_006009.3(TUBA1A):c.1264C>T (p.Arg422Cys)	TUBA1A	12	49185102	Lissencephaly 3;not provided
NM_006009.3(TUBA1A):c.1265G>A (p.Arg422His)	TUBA1A	12	49185101	Lissencephaly 3;not provided
NM_001143979.1(NDE1):c.658C>T (p.Arg220Ter)	NDE1	16	15691278	Lissencephaly 4
NM_005886.2(KATNB1):c.1604C>T (p.Ser535Leu)	KATNB1	16	57755878	Lissencephaly 6, with microcephaly
NM_005886.2(KATNB1):c.432+1G>A	KATNB1	16	57751303	Lissencephaly 6, with microcephaly
NM_004935.3(CDK5):c.580+1G>A	CDK5	7	151055276	Lissencephaly 7 with cerebellar hypoplasia
NM_181783.3(TMTC3):c.2617C>T (p.Gln873Ter)	TMTC3	12	88195521	LISSENCEPHALY 8
NM_181783.3(TMTC3):c.3G>A (p.Met1Ile)	TMTC3	12	88148318	LISSENCEPHALY 8
NM_178152.2(DCX):c.184G>A (p.Asp62Asn)	DCX	X	111410215	Lissencephaly, X-linked;Subcortical laminar heterotopia, X-linked
NM_018006.4(TRMU):c.815G>A (p.Gly272Asp)	TRMU	22	46353809	Liver failure acute infantile
NM_018006.4(TRMU):c.835G>A (p.Val279Met)	TRMU	22	46353829	Liver failure acute infantile;not provided
NM_004612.3(TGFBR1):c.599C>T (p.Thr200Ile)	TGFBR1	9	99137883	Loeys-Dietz syndrome 1
NM_003242.5(TGFBR2):c.1524G>A (p.Gln508=)	TGFBR2	3	30688511	Loeys-Dietz syndrome 2
NM_003242.5(TGFBR2):c.1346C>T (p.Ser449Phe)	TGFBR2	3	30674196	Loeys-Dietz syndrome 2
NM_003242.5(TGFBR2):c.1280C>T (p.Pro427Leu)	TGFBR2	3	30674130	Loeys-Dietz syndrome 2
NM_003242.5(TGFBR2):c.1609C>T (p.Arg537Cys)	TGFBR2	3	30691504	Loeys-Dietz syndrome 2;not provided
NM_003242.5(TGFBR2):c.1582C>T (p.Arg528Cys)	TGFBR2	3	30691477	Loeys-Dietz syndrome 2;not provided
NM_003242.5(TGFBR2):c.1379G>A (p.Arg460His)	TGFBR2	3	30674229	Loeys-Dietz syndrome 2;not provided
NM_003242.5(TGFBR2):c.1378C>T (p.Arg460Cys)	TGFBR2	3	30674228	Loeys-Dietz syndrome 2;Thoracic aortic aneurysm and aortic dissection;not provided
NM_005902.3(SMAD3):c.782C>T (p.Thr261Ile)	SMAD3	15	67181364	Loeys-Dietz syndrome 3
NM_005902.3(SMAD3):c.836G>A (p.Arg279Lys)	SMAD3	15	67181418	Loeys-Dietz syndrome 3
NM_005902.3(SMAD3):c.335C>T (p.Ala112Val)	SMAD3	15	67165023	Loeys-Dietz syndrome 3
NM_005902.3(SMAD3):c.788C>T (p.Pro263Leu)	SMAD3	15	67181370	Loeys-Dietz syndrome 3
NM_005902.3(SMAD3):c.859C>T (p.Arg287Trp)	SMAD3	15	67181441	Loeys-Dietz syndrome 3;not provided
NM_003239.4(TGFB3):c.899G>A (p.Arg300Gln)	TGFB3	14	75963343	Loeys-Dietz syndrome 4;Loeys-Dietz syndrome 5;not provided
NM_003238.3(TGFB2):c.895C>T (p.Arg299Trp)	TGFB2	1	218436110	Loeys-Dietz syndrome 4;not provided
NM_001135599.2(TGFB2):c.988C>T (p.Arg330Cys)	TGFB2	1	218436119	Loeys-Dietz syndrome 4;not provided
NM_003239.4(TGFB3):c.1226G>A (p.Cys409Tyr)	TGFB3	14	75959200	Loeys-Dietz syndrome 5
NM_003239.4(TGFB3):c.898C>T (p.Arg300Trp)	TGFB3	14	75963344	Loeys-Dietz syndrome 5;not provided
NM_004612.3(TGFBR1):c.722C>T (p.Ser241Leu)	TGFBR1	9	99138006	Loeys-Dietz syndrome;Loeys-Dietz syndrome 1;Thoracic aortic aneurysm and aortic dissection;not provided
NM_004612.3(TGFBR1):c.1460G>A (p.Arg487Gln)	TGFBR1	9	99149253	Loeys-Dietz syndrome;Loeys-Dietz syndrome 1;Thoracic aortic aneurysm and aortic dissection;not provided
NM_004612.3(TGFBR1):c.1459C>T (p.Arg487Trp)	TGFBR1	9	99149252	Loeys-Dietz syndrome;Loeys-Dietz syndrome 1;Thoracic aortic aneurysm and aortic dissection;not provided
NM_003242.5(TGFBR2):c.1483C>T (p.Arg495Ter)	TGFBR2	3	30688470	Loeys-Dietz syndrome;Loeys-Dietz syndrome 2;Thoracic aortic aneurysm and aortic dissection;not provided
NM_003242.5(TGFBR2):c.1570G>A (p.Asp524Asn)	TGFBR2	3	30691465	Loeys-Dietz syndrome;not provided
NM_000218.2(KCNQ1):c.1175G>A (p.Trp392Ter)	KCNQ1	11	2587616	Long QT syndrome
NM_000218.2(KCNQ1):c.557G>A (p.Gly186Asp)	KCNQ1	11	2570707	Long QT syndrome 1

NM_174934.3(SCN4B):c.535C>T (p.Leu179Phe)	SCN4B	11	118141265	Long QT syndrome 10
NM_005751.4(AKAP9):c.4709C>T (p.Ser1570Leu)	AKAP9	7	92040690	Long QT syndrome 11
NM_003098.2(SNTA1):c.1169C>T (p.Ala390Val)	SNTA1	20	33410203	Long QT syndrome 12;not provided
NM_032834.3(ALG10):c.1339G>A (p.Val447Ile)	ALG10	12	34026832	Long QT syndrome 2
NM_000238.3(KCNH2):c.3003G>A (p.Trp1001Ter)	KCNH2	7	150947477	Long QT syndrome 2;not provided
NM_033337.2(CAV3):c.253G>A (p.Ala85Thr)	CAV3	3	8745664	Long QT syndrome 9;not provided
NM_000218.2(KCNQ1):c.683+5G>A	KCNQ1	11	2571408	Long QT syndrome, LQT1 subtype;not provided
NM_000218.2(KCNQ1):c.1022C>T (p.Ala341Val)	KCNQ1	11	2583535	Long QT syndrome;Long QT syndrome 1;not provided
NM_000218.2(KCNQ1):c.1032G>A (p.Ala344=)	KCNQ1	11	2583545	Long QT syndrome;Long QT syndrome 1;not provided
NM_000238.3(KCNH2):c.2230C>T (p.Arg744Ter)	KCNH2	7	150950336	Long QT syndrome;Long QT syndrome 2;not provided
NM_000238.3(KCNH2):c.2587C>T (p.Arg863Ter)	KCNH2	7	150948861	Long QT syndrome;Long QT syndrome 2;not provided
NM_000218.2(KCNQ1):c.1032+1G>A	KCNQ1	11	2583546	Long QT syndrome;Long QT syndrome, LQT1 subtype;not provided
NM_000218.2(KCNQ1):c.1066C>T (p.Gln356Ter)	KCNQ1	11	2585245	Long QT syndrome;not provided
NM_000218.2(KCNQ1):c.1075C>T (p.Gln359Ter)	KCNQ1	11	2585254	Long QT syndrome;not provided
NM_000218.2(KCNQ1):c.1588C>T (p.Gln530Ter)	KCNQ1	11	2768917	Long QT syndrome;not provided
NM_000218.2(KCNQ1):c.477+5G>A	KCNQ1	11	2528023	Long QT syndrome;not provided
NM_000218.2(KCNQ1):c.914G>A (p.Trp305Ter)	KCNQ1	11	2572979	Long QT syndrome;not provided
NM_000719.6(CACNA1C):c.1552C>T (p.Arg518Cys)	CACNA1C	12	2566465	Long QT syndrome;not provided
NM_000238.3(KCNH2):c.3040C>T (p.Arg1014Ter)	KCNH2	7	150947440	Long QT syndrome;not provided
NM_000238.3(KCNH2):c.3002G>A (p.Trp1001Ter)	KCNH2	7	150947478	Long QT syndrome;not provided
NM_000218.2(KCNQ1):c.477+1G>A	KCNQ1	11	2528019	Long QT syndrome;not provided
NM_000238.3(KCNH2):c.2145+1G>A	KCNH2	7	150950920	Long QT syndrome;not provided
NM_033337.2(CAV3):c.277G>A (p.Ala93Thr)	CAV3	3	8745688	Long QT syndrome;Rippling muscle disease 2, autosomal recessive;not provided
NM_000182.4(HADHA):c.1918C>T (p.Gln640Ter)	HADHA	2	26192392	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency;Mitochondrial trifunctional protein deficiency
NM_000182.4(HADHA):c.2146+1G>A	HADHA	2	26191482	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency;Mitochondrial trifunctional protein deficiency
NM_000182.4(HADHA):c.157C>T (p.Arg53Ter)	HADHA	2	26238957	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency;Mitochondrial trifunctional protein deficiency;not provided
NM_002111.8(HTT):c.4469+1G>A	HTT	4	3177388	LOPES-MACIEL-RODAN SYNDROME
NM_000276.3(OCRL):c.2530C>T (p.Arg844Ter)	OCRL	X	129589905	Lowe syndrome
NM_000276.3(OCRL):c.1499G>A (p.Arg500Gln)	OCRL	X	129569296	Lowe syndrome
NM_000276.3(OCRL):c.1714-1G>A	OCRL	X	129575896	Lowe syndrome
NM_181699.2(PPP2R1B):c.269G>A (p.Gly90Asp)	PPP2R1B	11	111764842	Lung cancer
NM_183233.2(SLC22A18):c.698C>T (p.Ser233Phe)	SLC22A18	11	2918030	Lung cancer
NM_000745.3(CHRNA5):c.1192G>A (p.Asp398Asn)	CHRNA5	15	78590583	Lung cancer susceptibility 2;Smoking as a quantitative trait locus 3
NM_138704.3(NSMCE3):c.790C>T (p.Leu264Phe)	NSMCE3	15	29268916	LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME;Lung damage, immunodeficiency and chromosome breakage syndrome
NM_138704.3(NSMCE3):c.626C>T (p.Pro209Leu)	NSMCE3	15	29269080	LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME;Lung damage, immunodeficiency and chromosome breakage syndrome
NM_014159.6(SETD2):c.820C>T (p.Gln274Ter)	SETD2	3	47123816	Luscan-lumish syndrome
NM_000548.4(TSC2):c.5024C>T (p.Pro1675Leu)	TSC2	16	2087897	Lymphangiomyomatosis;Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.1513C>T (p.Arg505Ter)	TSC2	16	2064341	Lymphangiomyomatosis;Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.1832G>A (p.Arg611Gln)	TSC2	16	2070571	Lymphangiomyomatosis;Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.491G>A (p.Trp164Ter)	TSC1	9	132923365	Lymphangiomyomatosis;Tuberous sclerosis syndrome;not provided

NM_000548.4(TSC2):c.3442C>T (p.Gln1148Ter)	TSC2	16	2080209	Lymphangiomyomatosis;Tuberous sclerosis syndrome;not provided
NM_020435.3(GJC2):c.143C>T (p.Ser48Leu)	GJC2	1	228157901	Lymphedema, hereditary, IC
NM_020435.3(GJC2):c.778C>T (p.Arg260Cys)	GJC2	1	228158536	Lymphedema, hereditary, IC
NM_005429.4(VEGFC):c.628C>T (p.Arg210Ter)	VEGFC	4	176711575	Lymphedema, hereditary, ID
NM_001142864.3(PIEZO1):c.6682C>T (p.Gln2228Ter)	PIEZO1	16	88716877	Lymphedema, hereditary, III
NM_001142864.3(PIEZO1):c.3700_3796del97	PIEZO1	16	88726546	Lymphedema, hereditary, III
NM_001142864.3(PIEZO1):c.1669+1G>A	PIEZO1	16	88735134	Lymphedema, hereditary, III
NM_001142864.3(PIEZO1):c.7289C>T (p.Pro2430Leu)	PIEZO1	16	88715960	Lymphedema, hereditary, III
NM_001145661.1(GATA2):c.1009C>T (p.Arg337Ter)	GATA2	3	128483868	Lymphedema, primary, with myelodysplasia
NM_005546.3(ITK):c.1003C>T (p.Arg335Trp)	ITK	5	157241663	Lymphoproliferative syndrome 1
NM_005546.3(ITK):c.86G>A (p.Arg29His)	ITK	5	157181063	Lymphoproliferative syndrome 1
NM_002351.4(SH2D1A):c.163C>T (p.Arg55Ter)	SH2D1A	X	124365786	Lymphoproliferative syndrome 1, X-linked
NM_002351.4(SH2D1A):c.172C>T (p.Gln58Ter)	SH2D1A	X	124365795	Lymphoproliferative syndrome 1, X-linked
NM_002351.4(SH2D1A):c.302C>T (p.Pro101Leu)	SH2D1A	X	124370276	Lymphoproliferative syndrome 1, X-linked
NM_002351.4(SH2D1A):c.203C>T (p.Thr68Ile)	SH2D1A	X	124370177	Lymphoproliferative syndrome 1, X-linked
NM_001242.4(CD27):c.24G>A (p.Trp8Ter)	CD27	12	6445119	Lymphoproliferative syndrome 2
NM_001242.4(CD27):c.158G>A (p.Cys53Tyr)	CD27	12	6445445	Lymphoproliferative syndrome 2
NM_001167.3(XIAP):c.608G>A (p.Cys203Tyr)	XIAP	X	123886270	Lymphoproliferative syndrome 2, X-linked
NM_000249.3(MLH1):c.454-1G>A	MLH1	3	37008813	Lynch syndrome
NM_000251.2(MSH2):c.1030C>T (p.Gln344Ter)	MSH2	2	47416383	Lynch syndrome
NM_000179.2(MSH6):c.2330G>A (p.Trp777Ter)	MSH6	2	47800313	Lynch syndrome
NM_000179.2(MSH6):c.2191C>T (p.Gln731Ter)	MSH6	2	47800174	Lynch syndrome
NM_000179.2(MSH6):c.2764C>T (p.Arg922Ter)	MSH6	2	47800747	Lynch syndrome
NM_000179.2(MSH6):c.2815C>T (p.Gln939Ter)	MSH6	2	47800798	Lynch syndrome
NM_000179.2(MSH6):c.3020G>A (p.Trp1007Ter)	MSH6	2	47801003	Lynch syndrome
NM_000179.2(MSH6):c.3436C>T (p.Gln1146Ter)	MSH6	2	47803683	Lynch syndrome
NM_000179.2(MSH6):c.3647-1G>A	MSH6	2	47806203	Lynch syndrome
NM_000179.2(MSH6):c.3772C>T (p.Gln1258Ter)	MSH6	2	47806329	Lynch syndrome
NM_000179.2(MSH6):c.3838C>T (p.Gln1280Ter)	MSH6	2	47806488	Lynch syndrome
NM_000179.2(MSH6):c.706C>T (p.Gln236Ter)	MSH6	2	47798689	Lynch syndrome
NM_000179.2(MSH6):c.730C>T (p.Gln244Ter)	MSH6	2	47798713	Lynch syndrome
NM_000249.3(MLH1):c.1171C>T (p.Gln391Ter)	MLH1	3	37025769	Lynch syndrome
NM_000249.3(MLH1):c.1192C>T (p.Gln398Ter)	MLH1	3	37025790	Lynch syndrome
NM_000249.3(MLH1):c.1225C>T (p.Gln409Ter)	MLH1	3	37025823	Lynch syndrome
NM_000249.3(MLH1):c.1276C>T (p.Gln426Ter)	MLH1	3	37025874	Lynch syndrome
NM_000249.3(MLH1):c.1528C>T (p.Gln510Ter)	MLH1	3	37028902	Lynch syndrome
NM_000249.3(MLH1):c.1609C>T (p.Gln537Ter)	MLH1	3	37040236	Lynch syndrome
NM_000249.3(MLH1):c.1613G>A (p.Trp538Ter)	MLH1	3	37040240	Lynch syndrome
NM_000249.3(MLH1):c.1614G>A (p.Trp538Ter)	MLH1	3	37040241	Lynch syndrome
NM_000249.3(MLH1):c.1624C>T (p.Gln542Ter)	MLH1	3	37040251	Lynch syndrome
NM_000249.3(MLH1):c.1684C>T (p.Gln562Ter)	MLH1	3	37042284	Lynch syndrome
NM_000249.3(MLH1):c.1731+1G>A	MLH1	3	37042332	Lynch syndrome
NM_000249.3(MLH1):c.1731+5G>A	MLH1	3	37042336	Lynch syndrome
NM_000249.3(MLH1):c.1732-1G>A	MLH1	3	37047518	Lynch syndrome
NM_000249.3(MLH1):c.1896G>A (p.Glu632=)	MLH1	3	37047683	Lynch syndrome

NM_000249.3(MLH1):c.1989+1G>A	MLH1	3	37048610	Lynch syndrome
NM_000249.3(MLH1):c.1990-1G>A	MLH1	3	37048903	Lynch syndrome
NM_000249.3(MLH1):c.1998G>A (p.Trp666Ter)	MLH1	3	37048912	Lynch syndrome
NM_000249.3(MLH1):c.208-1G>A	MLH1	3	37000954	Lynch syndrome
NM_000249.3(MLH1):c.2101C>T (p.Gln701Ter)	MLH1	3	37049015	Lynch syndrome
NM_000249.3(MLH1):c.2136G>A (p.Trp712Ter)	MLH1	3	37050518	Lynch syndrome
NM_000249.3(MLH1):c.2224C>T (p.Gln742Ter)	MLH1	3	37050606	Lynch syndrome
NM_000249.3(MLH1):c.230G>A (p.Cys77Tyr)	MLH1	3	37000977	Lynch syndrome
NM_000249.3(MLH1):c.256C>T (p.Gln86Ter)	MLH1	3	37001003	Lynch syndrome
NM_000249.3(MLH1):c.436C>T (p.Gln146Ter)	MLH1	3	37007046	Lynch syndrome
NM_000249.3(MLH1):c.445C>T (p.Gln149Ter)	MLH1	3	37007055	Lynch syndrome
NM_000249.3(MLH1):c.545G>A (p.Arg182Lys)	MLH1	3	37008905	Lynch syndrome
NM_000249.3(MLH1):c.731G>A (p.Gly244Asp)	MLH1	3	37014485	Lynch syndrome
NM_000249.3(MLH1):c.76C>T (p.Gln26Ter)	MLH1	3	36993623	Lynch syndrome
NM_000249.3(MLH1):c.842C>T (p.Ala281Val)	MLH1	3	37017557	Lynch syndrome
NM_000249.3(MLH1):c.882C>T (p.Leu294=)	MLH1	3	37017597	Lynch syndrome
NM_000249.3(MLH1):c.901C>T (p.Gln301Ter)	MLH1	3	37020326	Lynch syndrome
NM_000251.2(MSH2):c.1013G>A (p.Gly338Glu)	MSH2	2	47416366	Lynch syndrome
NM_000251.2(MSH2):c.1034G>A (p.Trp345Ter)	MSH2	2	47416387	Lynch syndrome
NM_000251.2(MSH2):c.1129C>T (p.Gln377Ter)	MSH2	2	47429794	Lynch syndrome
NM_000251.2(MSH2):c.1183C>T (p.Gln395Ter)	MSH2	2	47429848	Lynch syndrome
NM_000251.2(MSH2):c.1189C>T (p.Gln397Ter)	MSH2	2	47429854	Lynch syndrome
NM_000251.2(MSH2):c.1204C>T (p.Gln402Ter)	MSH2	2	47429869	Lynch syndrome
NM_000251.2(MSH2):c.1276+1G>A	MSH2	2	47429942	Lynch syndrome
NM_000251.2(MSH2):c.1528C>T (p.Gln510Ter)	MSH2	2	47466675	Lynch syndrome
NM_000251.2(MSH2):c.1552C>T (p.Gln518Ter)	MSH2	2	47466699	Lynch syndrome
NM_000251.2(MSH2):c.1720C>T (p.Gln574Ter)	MSH2	2	47471023	Lynch syndrome
NM_000251.2(MSH2):c.1777C>T (p.Gln593Ter)	MSH2	2	47475042	Lynch syndrome
NM_000251.2(MSH2):c.1885C>T (p.Gln629Ter)	MSH2	2	47475150	Lynch syndrome
NM_000251.2(MSH2):c.2087C>T (p.Pro696Leu)	MSH2	2	47476448	Lynch syndrome
NM_000251.2(MSH2):c.2251G>A (p.Gly751Arg)	MSH2	2	47478312	Lynch syndrome
NM_000251.2(MSH2):c.2291G>A (p.Trp764Ter)	MSH2	2	47478352	Lynch syndrome
NM_000251.2(MSH2):c.2292G>A (p.Trp764Ter)	MSH2	2	47478353	Lynch syndrome
NM_000251.2(MSH2):c.2446C>T (p.Gln816Ter)	MSH2	2	47478507	Lynch syndrome
NM_000251.2(MSH2):c.2470C>T (p.Gln824Ter)	MSH2	2	47480707	Lynch syndrome
NM_000251.2(MSH2):c.2536C>T (p.Gln846Ter)	MSH2	2	47480773	Lynch syndrome
NM_000251.2(MSH2):c.2581C>T (p.Gln861Ter)	MSH2	2	47480818	Lynch syndrome
NM_000251.2(MSH2):c.2634G>A (p.Glu878=)	MSH2	2	47480871	Lynch syndrome
NM_000251.2(MSH2):c.2635C>T (p.Gln879Ter)	MSH2	2	47482779	Lynch syndrome
NM_000251.2(MSH2):c.28C>T (p.Gln10Ter)	MSH2	2	47403219	Lynch syndrome
NM_000251.2(MSH2):c.472C>T (p.Gln158Ter)	MSH2	2	47410199	Lynch syndrome
NM_000251.2(MSH2):c.478C>T (p.Gln160Ter)	MSH2	2	47410205	Lynch syndrome
NM_000251.2(MSH2):c.484G>A (p.Gly162Arg)	MSH2	2	47410211	Lynch syndrome
NM_000251.2(MSH2):c.490G>A (p.Gly164Arg)	MSH2	2	47410217	Lynch syndrome
NM_000251.2(MSH2):c.547C>T (p.Gln183Ter)	MSH2	2	47410274	Lynch syndrome

NM_000251.2(MSH2):c.577C>T (p.Gln193Ter)	MSH2	2	47410304	Lynch syndrome
NM_000251.2(MSH2):c.643C>T (p.Gln215Ter)	MSH2	2	47410370	Lynch syndrome
NM_000251.2(MSH2):c.645+1G>A	MSH2	2	47410373	Lynch syndrome
NM_000251.2(MSH2):c.652C>T (p.Gln218Ter)	MSH2	2	47412420	Lynch syndrome
NM_000251.2(MSH2):c.754C>T (p.Gln252Ter)	MSH2	2	47412522	Lynch syndrome
NM_000251.2(MSH2):c.792+1G>A	MSH2	2	47412561	Lynch syndrome
NM_000251.2(MSH2):c.942G>A (p.Gln314=)	MSH2	2	47414418	Lynch syndrome
NM_000535.6(PMS2):c.949C>T (p.Gln317Ter)	PMS2	7	5992012	Lynch syndrome
NM_000179.2(MSH6):c.1367G>A (p.Trp456Ter)	MSH6	2	47799350	Lynch syndrome
NM_000179.2(MSH6):c.1045C>T (p.Gln349Ter)	MSH6	2	47799028	Lynch syndrome
NM_000251.2(MSH2):c.1384C>T (p.Gln462Ter)	MSH2	2	47445655	Lynch syndrome
NM_002354.2(EPCAM):c.133C>T (p.Gln45Ter)	EPCAM	2	47373519	Lynch syndrome
NM_002354.2(EPCAM):c.429G>A (p.Trp143Ter)	EPCAM	2	47375237	Lynch syndrome
NM_002354.2(EPCAM):c.523C>T (p.Gln175Ter)	EPCAM	2	47377045	Lynch syndrome
NM_000179.2(MSH6):c.2680C>T (p.Gln894Ter)	MSH6	2	47800663	Lynch syndrome
NM_000251.2(MSH2):c.350G>A (p.Trp117Ter)	MSH2	2	47408539	Lynch syndrome
NM_000179.2(MSH6):c.2735G>A (p.Trp912Ter)	MSH6	2	47800718	Lynch syndrome
NM_000179.2(MSH6):c.3556+1G>A	MSH6	2	47805028	Lynch syndrome
NM_000251.2(MSH2):c.388C>T (p.Gln130Ter)	MSH2	2	47410115	Lynch syndrome
NM_000535.6(PMS2):c.1912C>T (p.Gln638Ter)	PMS2	7	5986853	Lynch syndrome
NM_000535.6(PMS2):c.1891C>T (p.Gln631Ter)	PMS2	7	5986874	Lynch syndrome
NM_000249.3(MLH1):c.306+1G>A	MLH1	3	37001054	Lynch syndrome I
NM_000249.3(MLH1):c.62C>T (p.Ala21Val)	MLH1	3	36993609	Lynch syndrome I
NM_000251.2(MSH2):c.1865C>T (p.Pro622Leu)	MSH2	2	47475130	Lynch syndrome;Lynch syndrome I
NM_000179.2(MSH6):c.426G>A (p.Trp142Ter)	MSH6	2	47791092	Lynch syndrome;Lynch syndrome I
NM_000251.2(MSH2):c.715C>T (p.Gln239Ter)	MSH2	2	47412483	Lynch syndrome;Lynch syndrome I
NM_000249.3(MLH1):c.350C>T (p.Thr117Met)	MLH1	3	37004444	Lynch syndrome;Lynch syndrome II;Lynch syndrome II;not provided
NM_000251.2(MSH2):c.1915C>T (p.His639Tyr)	MSH2	2	47475180	Lynch syndrome;Lynch syndrome I;not provided
NM_000251.2(MSH2):c.289C>T (p.Gln97Ter)	MSH2	2	47408478	Lynch syndrome;Lynch syndrome I;not provided
NM_000251.2(MSH2):c.2785C>T (p.Arg929Ter)	MSH2	2	47482929	Lynch syndrome;Lynch syndrome I;not specified
NM_000249.3(MLH1):c.131C>T (p.Ser44Phe)	MLH1	3	36996633	Lynch syndrome;Lynch syndrome II
NM_000249.3(MLH1):c.306+5G>A	MLH1	3	37001058	Lynch syndrome;Lynch syndrome II;not provided
NM_000249.3(MLH1):c.1219C>T (p.Gln407Ter)	MLH1	3	37025817	Lynch syndrome;Lynch syndrome II;not provided
NM_000251.2(MSH2):c.1801C>T (p.Gln601Ter)	MSH2	2	47475066	Lynch syndrome;MuIr-TorrÃ© syndrome;not provided
NM_000249.3(MLH1):c.298C>T (p.Arg100Ter)	MLH1	3	37001045	Lynch syndrome;not provided
NM_000179.2(MSH6):c.3013C>T (p.Arg1005Ter)	MSH6	2	47800996	Lynch syndrome;not provided
NM_000179.2(MSH6):c.694C>T (p.Gln232Ter)	MSH6	2	47798677	Lynch syndrome;not provided
NM_000179.2(MSH6):c.742C>T (p.Arg248Ter)	MSH6	2	47798725	Lynch syndrome;not provided
NM_000249.3(MLH1):c.1039-1G>A	MLH1	3	37025636	Lynch syndrome;not provided
NM_000249.3(MLH1):c.142C>T (p.Gln48Ter)	MLH1	3	36996644	Lynch syndrome;not provided
NM_000249.3(MLH1):c.1790G>A (p.Trp597Ter)	MLH1	3	37047577	Lynch syndrome;not provided
NM_000249.3(MLH1):c.1961C>T (p.Pro654Leu)	MLH1	3	37048581	Lynch syndrome;not provided
NM_000249.3(MLH1):c.2103+1G>A	MLH1	3	37049018	Lynch syndrome;not provided
NM_000249.3(MLH1):c.2135G>A (p.Trp712Ter)	MLH1	3	37050517	Lynch syndrome;not provided
NM_000249.3(MLH1):c.588+5G>A	MLH1	3	37011867	Lynch syndrome;not provided

NM_000249.3(MLH1):c.790+1G>A	MLH1	3	37014545	Lynch syndrome;not provided
NM_000251.2(MSH2):c.1035G>A (p.Trp345Ter)	MSH2	2	47416388	Lynch syndrome;not provided
NM_000251.2(MSH2):c.1255C>T (p.Gln419Ter)	MSH2	2	47429920	Lynch syndrome;not provided
NM_000251.2(MSH2):c.1861C>T (p.Arg621Ter)	MSH2	2	47475126	Lynch syndrome;not provided
NM_000251.2(MSH2):c.226C>T (p.Gln76Ter)	MSH2	2	47408415	Lynch syndrome;not provided
NM_000251.2(MSH2):c.2653C>T (p.Gln885Ter)	MSH2	2	47482797	Lynch syndrome;not provided
NM_000251.2(MSH2):c.508C>T (p.Gln170Ter)	MSH2	2	47410235	Lynch syndrome;not provided
NM_000251.2(MSH2):c.862C>T (p.Gln288Ter)	MSH2	2	47414338	Lynch syndrome;not provided
NM_000251.2(MSH2):c.892C>T (p.Gln298Ter)	MSH2	2	47414368	Lynch syndrome;not provided
NM_000251.2(MSH2):c.970C>T (p.Gln324Ter)	MSH2	2	47416323	Lynch syndrome;not provided
NM_000535.6(PMS2):c.1144+1G>A	PMS2	7	5989799	Lynch syndrome;not provided
NM_000251.2(MSH2):c.1984C>T (p.Gln662Ter)	MSH2	2	47475249	Lynch syndrome;not provided
NM_000249.3(MLH1):c.381-1G>A	MLH1	3	37006990	Lynch syndrome;not provided
NM_000535.6(PMS2):c.631C>T (p.Arg211Ter)	PMS2	7	5999182	Lynch syndrome;not provided
NM_000251.2(MSH2):c.790C>T (p.Gln264Ter)	MSH2	2	47412558	Lynch syndrome;not provided
NM_000251.2(MSH2):c.366+1G>A	MSH2	2	47408556	Lynch syndrome;not provided
NM_000179.2(MSH6):c.4001G>A (p.Arg1334Gln)	MSH6	2	47806651	Lynch syndrome;not provided;not specified
NM_000251.2(MSH2):c.1662-1G>A	MSH2	2	47470964	Lynch syndrome;Turcot syndrome
NM_001126105.2(SLC7A7):c.1228C>T (p.Arg410Ter)	SLC7A7	14	22774371	Lysinuric protein intolerance
NM_001126105.2(SLC7A7):c.726G>A (p.Trp242Ter)	SLC7A7	14	22778837	Lysinuric protein intolerance
NM_000235.3(LIPA):c.253C>T (p.Gln85Ter)	LIPA	10	89228375	Lysosomal acid lipase deficiency
NM_000235.3(LIPA):c.894G>A (p.Gln298=)	LIPA	10	89222511	Lysosomal acid lipase deficiency;not provided
NM_032822.2(FAM136A):c.226C>T (p.Gln76Ter)	FAM136A	2	70300842	MÃ©niÃ¨re's disease
NM_001025.4(RPS23):c.200G>A (p.Arg67Lys)	RPS23	5	82276483	MACINNES SYNDROME
NM_003922.3(HERC1):c.2625G>A (p.Trp875Ter)	HERC1	15	63734745	Macrocephaly, dysmorphic facies, and psychomotor retardation
NM_003922.3(HERC1):c.13559G>A (p.Gly4520Glu)	HERC1	15	63623777	Macrocephaly, dysmorphic facies, and psychomotor retardation
NM_003922.3(HERC1):c.9748C>T (p.Arg3250Ter)	HERC1	15	63656210	Macrocephaly, dysmorphic facies, and psychomotor retardation;Megalencephaly with thick corpus callosum, cerebellar atrophy, and intellectual disability
NM_032322.3(RNF135):c.727C>T (p.Gln243Ter)	RNF135	17	30997289	Macrocephaly, macrosomia, facial dysmorphism syndrome
NM_032322.3(RNF135):c.857G>A (p.Arg286His)	RNF135	17	30998749	Macrocephaly, macrosomia, facial dysmorphism syndrome
NM_000314.6(PTEN):c.392C>T (p.Thr131Ile)	PTEN	10	87933151	Macrocephaly/autism syndrome
NM_030773.3(TUBB1):c.952C>T (p.Arg318Trp)	TUBB1	20	59024379	MACROTHROMBOCYTOPENIA, AUTOSOMAL DOMINANT, TUBB1-RELATED
NM_021615.4(CHST6):c.892C>T (p.Gln298Ter)	CHST6	16	75478937	Macular corneal dystrophy Type I
NM_001737.4(C9):c.499C>T (p.Pro167Ser)	C9	5	39331792	Macular degeneration, age-related, 15
NM_001903.4(CTNNA1):c.919G>A (p.Glu307Lys)	CTNNA1	5	138827575	Macular dystrophy, patterned, 2
NM_001563.3(IMPG1):c.1519C>T (p.Arg507Ter)	IMPG1	6	75950867	Macular dystrophy, vitelliform, 4
NM_000322.4(PRPH2):c.947G>A (p.Trp316Ter)	PRPH2	6	42698389	Macular dystrophy, vitelliform, adult-onset;not provided
NM_014646.2(LPIN2):c.2201C>T (p.Ser734Leu)	LPIN2	18	2922173	Majeed syndrome;not provided
NM_003921.4(BCL10):c.172C>T (p.Arg58Ter)	BCL10	1	85270792	Male germ cell tumor, somatic
NM_000069.2(CACNA1S):c.3257G>A (p.Arg1086His)	CACNA1S	1	201060815	Malignant hyperthermia susceptibility type 5
NM_000540.2(RYR1):c.1840C>T (p.Arg614Cys)	RYR1	19	38457545	Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.742G>A (p.Gly248Arg)	RYR1	19	38446710	Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.7300G>A (p.Gly2434Arg)	RYR1	19	38499993	Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.7372C>T (p.Arg2458Cys)	RYR1	19	38500654	Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.6502G>A (p.Val2168Met)	RYR1	19	38494579	Malignant hyperthermia, susceptibility to, 1;not provided

NM_000540.2(RYR1):c.14477C>T (p.Thr4826Ile)	RYR1	19	38580094	Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.1589G>A (p.Arg530His)	RYR1	19	38455463	Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.5183C>T (p.Ser1728Phe)	RYR1	19	38485838	Malignant hyperthermia, susceptibility to, 1;not provided
NM_000540.2(RYR1):c.7048G>A (p.Ala2350Thr)	RYR1	19	38499655	Malignant hyperthermia, susceptibility to, 1;not provided
NM_032977.3(CASP10):c.1241C>T (p.Ala414Val)	CASP10	2	201209388	Malignant lymphoma, non-Hodgkin
NM_032977.3(CASP10):c.769C>T (p.Gln257Ter)	CASP10	2	201205929	Malignant lymphoma, non-Hodgkin;Neoplasm of stomach
NM_000314.6(PTEN):c.55G>A (p.Asp19Asn)	PTEN	10	87864524	Malignant melanoma
NM_000314.6(PTEN):c.649G>A (p.Val217Ile)	PTEN	10	87957867	Malignant melanoma
NM_002755.3(MAP2K1):c.607G>A (p.Glu203Lys)	MAP2K1	15	66481793	Malignant melanoma
NM_002755.3(MAP2K1):c.790C>T (p.Pro264Ser)	MAP2K1	15	66485086	Malignant melanoma
NM_004333.4(BRAF):c.1796C>T (p.Thr599Ile)	BRAF	7	140753339	Malignant melanoma;Neoplasm;not provided
NM_004333.4(BRAF):c.1798G>A (p.Val600Met)	BRAF	7	140753337	Malignant melanoma;Non-small cell lung cancer
NM_004333.4(BRAF):c.1390G>A (p.Gly464Arg)	BRAF	7	140781618	Malignant melanoma;not provided
NM_014225.5(PPP2R1A):c.536C>T (p.Pro179Leu)	PPP2R1A	19	52212718	Malignant neoplasm of body of uterus;Mental retardation, autosomal dominant 36;Uterine Carcinosarcoma;not provided
NM_000044.4(AR):c.1943G>A (p.Ser648Asn)	AR	X	67711459	Malignant tumor of prostate
NM_000044.4(AR):c.2191G>A (p.Val731Met)	AR	X	67717495	Malignant tumor of prostate
NM_000044.4(AR):c.2623C>T (p.His875Tyr)	AR	X	67723701	Malignant tumor of prostate
NM_000044.4(AR):c.2164G>A (p.Ala722Thr)	AR	X	67711680	Malignant tumor of prostate
NM_016218.3(POLK):c.85G>A (p.Glu29Lys)	POLK	5	75547107	Malignant tumor of prostate
NM_016218.3(POLK):c.410C>T (p.Ser137Phe)	POLK	5	75573739	Malignant tumor of prostate
NM_016218.3(POLK):c.461G>A (p.Gly154Glu)	POLK	5	75573790	Malignant tumor of prostate
NM_016218.3(POLK):c.1284G>A (p.Ala428=)	POLK	5	75590368	Malignant tumor of prostate
NM_016218.3(POLK):c.1324C>T (p.Leu442Phe)	POLK	5	75590408	Malignant tumor of prostate
NM_016218.3(POLK):c.1341G>A (p.Gln447=)	POLK	5	75590425	Malignant tumor of prostate
NM_016218.3(POLK):c.1345G>A (p.Glu449Lys)	POLK	5	75590429	Malignant tumor of prostate
NM_016218.3(POLK):c.1692G>A (p.Lys564=)	POLK	5	75596385	Malignant tumor of prostate
NM_016218.3(POLK):c.1741G>A (p.Asp581Asn)	POLK	5	75596434	Malignant tumor of prostate
NM_016218.3(POLK):c.2033C>T (p.Ser678Phe)	POLK	5	75596726	Malignant tumor of prostate
NM_000455.4(STK11):c.488G>A (p.Gly163Asp)	STK11	19	1220396	Malignant tumor of testis
NM_003921.4(BCL10):c.488C>T (p.Thr163Met)	BCL10	1	85267841	Malignant tumor of testis
NM_000368.4(TSC1):c.663+1G>A	TSC1	9	132921818	Malignant tumor of urinary bladder;not provided
NM_000368.4(TSC1):c.664-1G>A	TSC1	9	132921437	Malignant tumor of urinary bladder;Tuberous sclerosis 1
NM_000368.4(TSC1):c.1525C>T (p.Arg509Ter)	TSC1	9	132906053	Malignant tumor of urinary bladder;Tuberous sclerosis 1;Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.2074C>T (p.Arg692Ter)	TSC1	9	132903785	Malignant tumor of urinary bladder;Tuberous sclerosis 1;Tuberous sclerosis syndrome;not provided
NM_006438.4(COLEC10):c.25C>T (p.Arg9Ter)	COLEC10	8	119067306	Malpuech facial clefting syndrome
NM_170707.3(LMNA):c.1586C>T (p.Ala529Val)	LMNA	1	156137210	Mandibuloacral dysostosis;not provided
NM_170707.3(LMNA):c.1585G>A (p.Ala529Thr)	LMNA	1	156137209	Mandibuloacral dysostosis;not provided
NM_170707.3(LMNA):c.1620G>A (p.Met540Ile)	LMNA	1	156137665	Mandibuloacral dysplasia with type A lipodystrophy
NM_170707.3(LMNA):c.1318G>A (p.Val440Met)	LMNA	1	156136374	Mandibuloacral dysplasia with type A lipodystrophy, atypical;not provided
NM_005857.4(ZMPSTE24):c.121C>T (p.Gln41Ter)	ZMPSTE24	1	40258392	Mandibuloacral dysplasia with type B lipodystrophy;not provided
NM_005857.4(ZMPSTE24):c.743C>T (p.Pro248Leu)	ZMPSTE24	1	40272009	Mandibuloacral dysplasia with type B lipodystrophy;not provided
NM_005857.4(ZMPSTE24):c.1349G>A (p.Trp450Ter)	ZMPSTE24	1	40292590	Mandibuloacral dysplasia with type B lipodystrophy;not provided
NM_001957.3(EDNRA):c.907G>A (p.Glu303Lys)	EDNRA	4	147539823	Mandibulofacial dysostosis with alopecia
NM_000350.2(ABCA4):c.4139C>T (p.Pro1380Leu)	ABCA4	1	94031110	Mandibulofacial dysostosis with mental deficiency;Stargardt disease 1;not provided

NM_203290.3(POLR1C):c.835C>T (p.Arg279Trp)	POLR1C	6	43520961	Mandibulofacial dysostosis, Treacher Collins type, autosomal recessive;not provided
NM_203290.3(POLR1C):c.836G>A (p.Arg279Gln)	POLR1C	6	43520962	Mandibulofacial dysostosis, Treacher Collins type, autosomal recessive;Treacher Collins Syndrome, Recessive
NM_000242.2(MBL2):c.161G>A (p.Gly54Asp)	MBL2	10	52771475	Mannose-binding protein deficiency
NM_000242.2(MBL2):c.154C>T (p.Arg52Cys)	MBL2	10	52771482	Mannose-binding protein deficiency
NM_000051.3(ATM):c.4081C>T (p.Gln1361Ter)	ATM	11	108287687	Mantle cell lymphoma
NM_000709.3(BCKDHA):c.476G>A (p.Arg159Gln)	BCKDHA	19	41414149	Maple syrup urine disease
NM_183050.3(BCKDHB):c.3G>A (p.Met1Ile)	BCKDHB	6	80106696	Maple syrup urine disease
NM_183050.3(BCKDHB):c.554C>T (p.Pro185Leu)	BCKDHB	6	80168951	Maple syrup urine disease
NM_001918.3(DBT):c.1033G>A (p.Gly345Arg)	DBT	1	100206621	Maple syrup urine disease
NM_000709.3(BCKDHA):c.940C>T (p.Arg314Ter)	BCKDHA	19	41422715	Maple syrup urine disease
NM_000709.3(BCKDHA):c.745G>A (p.Gly249Ser)	BCKDHA	19	41422262	Maple syrup urine disease type 1A
NM_000709.3(BCKDHA):c.793C>T (p.Arg265Trp)	BCKDHA	19	41422310	Maple syrup urine disease type 1A;not provided
NM_000056.4(BCKDHB):c.616C>T (p.His206Tyr)	BCKDHB	6	80169013	MAPLE SYRUP URINE DISEASE, CLASSIC, TYPE IB
NM_000709.3(BCKDHA):c.868G>A (p.Gly290Arg)	BCKDHA	19	41422643	MAPLE SYRUP URINE DISEASE, INTERMEDIATE, TYPE IA;Maple syrup urine disease;not provided
NM_000108.4(DLD):c.1463C>T (p.Pro488Leu)	DLD	7	107919098	Maple syrup urine disease, type 3
NM_000108.4(DLD):c.1123G>A (p.Glu375Lys)	DLD	7	107917349	Maple syrup urine disease, type 3;not provided
NM_183050.3(BCKDHB):c.832G>A (p.Gly278Ser)	BCKDHB	6	80201023	Maple syrup urine disease;not provided
NM_000709.3(BCKDHA):c.1036C>T (p.Arg346Cys)	BCKDHA	19	41423038	Maple syrup urine disease;not provided
NM_000709.3(BCKDHA):c.1234G>A (p.Val412Met)	BCKDHA	19	41424504	Maple syrup urine disease;not provided
NM_000709.3(BCKDHA):c.288+1G>A	BCKDHA	19	41410817	Maple syrup urine disease;not provided
NM_000709.3(BCKDHA):c.288+9C>T	BCKDHA	19	41410825	Maple syrup urine disease;not provided
NM_000709.3(BCKDHA):c.632C>T (p.Thr211Met)	BCKDHA	19	41419282	Maple syrup urine disease;not provided
NM_000709.3(BCKDHA):c.659C>T (p.Ala220Val)	BCKDHA	19	41422176	Maple syrup urine disease;not provided
NM_000709.3(BCKDHA):c.964C>T (p.Gln322Ter)	BCKDHA	19	41422739	Maple syrup urine disease;not provided
NM_000709.3(BCKDHA):c.979G>A (p.Glu327Lys)	BCKDHA	19	41422754	Maple syrup urine disease;not provided
NM_001918.3(DBT):c.1291C>T (p.Arg431Ter)	DBT	1	100196413	Maple syrup urine disease;not provided
NM_001918.3(DBT):c.251G>A (p.Trp84Ter)	DBT	1	100235436	Maple syrup urine disease;not provided
NM_001918.3(DBT):c.871C>T (p.Arg291Ter)	DBT	1	100214885	Maple syrup urine disease;not provided
NM_001918.3(DBT):c.901C>T (p.Arg301Cys)	DBT	1	100214855	Maple syrup urine disease;not provided
NM_000056.4(BCKDHB):c.1016C>T (p.Ser339Leu)	BCKDHB	6	80273199	Maple syrup urine disease;not provided
NM_000056.4(BCKDHB):c.344-1G>A	BCKDHB	6	80167677	Maple syrup urine disease;not provided
NM_183050.3(BCKDHB):c.509G>A (p.Arg170His)	BCKDHB	6	80168906	Maple syrup urine disease;not provided
NM_000056.4(BCKDHB):c.633+1G>A	BCKDHB	6	80169031	Maple syrup urine disease;not provided
NM_183050.3(BCKDHB):c.799C>T (p.Gln267Ter)	BCKDHB	6	80200990	Maple syrup urine disease;not provided
NM_183050.3(BCKDHB):c.853C>T (p.Arg285Ter)	BCKDHB	6	80203114	Maple syrup urine disease;not provided
NM_000056.4(BCKDHB):c.952-1G>A	BCKDHB	6	80273134	Maple syrup urine disease;not provided
NM_183050.3(BCKDHB):c.970C>T (p.Arg324Ter)	BCKDHB	6	80273153	Maple syrup urine disease;not provided
NM_153334.6(SCARF2):c.773G>A (p.Cys258Tyr)	SCARF2	22	20431099	Marden Walker like syndrome
NM_022068.3(PIEZO2):c.8056C>T (p.Arg2686Cys)	PIEZO2	18	10671730	Marden-Walker syndrome
NM_000138.4(FBN1):c.8226+1G>A	FBN1	15	48412568	Marfan lipodystrophy syndrome
NM_000138.4(FBN1):c.8268G>A (p.Trp2756Ter)	FBN1	15	48411338	Marfan syndrome
NM_000138.4(FBN1):c.3350G>A (p.Cys1117Tyr)	FBN1	15	48487425	Marfan syndrome
NM_000138.4(FBN1):c.3725G>A (p.Cys1242Tyr)	FBN1	15	48483931	Marfan syndrome
NM_000138.4(FBN1):c.3386G>A (p.Cys1129Tyr)	FBN1	15	48487389	Marfan syndrome

NM_000138.4(FBN1):c.3662G>A (p.Cys1221Tyr)	FBN1	15	48485424	Marfan syndrome
NM_000138.4(FBN1):c.1051C>T (p.Gln351Ter)	FBN1	15	48520755	Marfan syndrome
NM_000138.4(FBN1):c.184C>T (p.Arg62Cys)	FBN1	15	48613073	Marfan syndrome
NM_000138.4(FBN1):c.2855-1G>A	FBN1	15	48490079	Marfan syndrome
NM_000138.4(FBN1):c.3164G>A (p.Cys1055Tyr)	FBN1	15	48488412	Marfan syndrome
NM_000138.4(FBN1):c.368G>A (p.Cys123Tyr)	FBN1	15	48600213	Marfan syndrome
NM_000138.4(FBN1):c.4955G>A (p.Cys1652Tyr)	FBN1	15	48464009	Marfan syndrome
NM_000138.4(FBN1):c.7180C>T (p.Arg2394Ter)	FBN1	15	48427591	Marfan syndrome
NM_000138.4(FBN1):c.8267G>A (p.Trp2756Ter)	FBN1	15	48411339	Marfan syndrome
NM_000428.2(LTBP2):c.1642C>T (p.Arg548Ter)	LTBP2	14	74551108	Marfan syndrome
NM_000138.4(FBN1):c.1496G>A (p.Cys499Tyr)	FBN1	15	48513641	Marfan syndrome
NM_000138.4(FBN1):c.1879C>T (p.Arg627Cys)	FBN1	15	48505106	Marfan syndrome
NM_000138.4(FBN1):c.6886C>T (p.Gln2296Ter)	FBN1	15	48428457	Marfan syndrome
NM_000138.4(FBN1):c.3373C>T (p.Arg1125Ter)	FBN1	15	48487402	Marfan syndrome
NM_000138.4(FBN1):c.640G>A (p.Gly214Ser)	FBN1	15	48537707	Marfan syndrome
NM_000138.4(FBN1):c.5038C>T (p.Gln1680Ter)	FBN1	15	48463926	Marfan syndrome
NM_000138.4(FBN1):c.434G>A (p.Cys145Tyr)	FBN1	15	48600147	Marfan syndrome
NM_000138.4(FBN1):c.2563C>T (p.Gln855Ter)	FBN1	15	48495237	Marfan syndrome
NM_000138.4(FBN1):c.7466G>A (p.Cys2489Tyr)	FBN1	15	48422056	Marfan syndrome
NM_000138.4(FBN1):c.2089C>T (p.Gln697Ter)	FBN1	15	48503811	Marfan syndrome
NM_000138.4(FBN1):c.592C>T (p.Gln198Ter)	FBN1	15	48537755	Marfan syndrome
NM_000138.4(FBN1):c.6695G>A (p.Cys2232Tyr)	FBN1	15	48432910	Marfan syndrome
NM_000138.4(FBN1):c.6164-1G>A	FBN1	15	48437918	Marfan syndrome
NM_000138.4(FBN1):c.5627G>A (p.Cys1876Tyr)	FBN1	15	48448812	Marfan syndrome
NM_000138.4(FBN1):c.4061G>A (p.Trp1354Ter)	FBN1	15	48474554	Marfan syndrome
NM_000138.4(FBN1):c.1982G>A (p.Cys661Tyr)	FBN1	15	48503918	Marfan syndrome
NM_000138.4(FBN1):c.6784C>T (p.Gln2262Ter)	FBN1	15	48430758	Marfan syndrome
NM_000138.4(FBN1):c.409C>T (p.Gln137Ter)	FBN1	15	48600172	Marfan syndrome
NM_000138.4(FBN1):c.2954G>A (p.Gly985Glu)	FBN1	15	48489979	Marfan syndrome, atypical
NM_000138.4(FBN1):c.4087+1G>A	FBN1	15	48474527	Marfan syndrome, neonatal
NM_000138.4(FBN1):c.3095G>A (p.Cys1032Tyr)	FBN1	15	48488481	Marfan syndrome, neonatal
NM_000138.4(FBN1):c.3257G>A (p.Cys1086Tyr)	FBN1	15	48488193	Marfan syndrome, neonatal
NM_000138.4(FBN1):c.364C>T (p.Arg122Cys)	FBN1	15	48600217	Marfan syndrome;Marfan syndrome, atypical;not provided
NM_000138.4(FBN1):c.7726C>T (p.Arg2576Cys)	FBN1	15	48420780	Marfan syndrome;Marfan syndrome, autosomal recessive
NM_000138.4(FBN1):c.3217G>A (p.Glu1073Lys)	FBN1	15	48488233	Marfan syndrome;Marfan syndrome, neonatal;not provided
NM_000138.4(FBN1):c.1585C>T (p.Arg529Ter)	FBN1	15	48513552	Marfan syndrome;not provided
NM_000138.4(FBN1):c.4460-8G>A	FBN1	15	48468542	Marfan syndrome;not provided
NM_000138.4(FBN1):c.4786C>T (p.Arg1596Ter)	FBN1	15	48465820	Marfan syndrome;not provided
NM_000138.4(FBN1):c.7806G>A (p.Trp2602Ter)	FBN1	15	48420700	Marfan syndrome;not provided
NM_000138.4(FBN1):c.247+1G>A	FBN1	15	48613009	Marfan syndrome;not provided
NM_000138.4(FBN1):c.2495G>A (p.Cys832Tyr)	FBN1	15	48495513	Marfan syndrome;not provided
NM_000138.4(FBN1):c.493C>T (p.Arg165Ter)	FBN1	15	48596328	Marfan syndrome;not provided
NM_000138.4(FBN1):c.5504G>A (p.Cys1835Tyr)	FBN1	15	48452603	Marfan syndrome;not provided
NM_000138.4(FBN1):c.5863C>T (p.Gln1955Ter)	FBN1	15	48445430	Marfan syndrome;not provided
NM_000138.4(FBN1):c.6658C>T (p.Arg2220Ter)	FBN1	15	48432947	Marfan syndrome;not provided

NM_000138.4(FBN1):c.7606G>A (p.Gly2536Arg)	FBN1	15	48421651	Marfan syndrome;not provided
NM_000138.4(FBN1):c.7955G>A (p.Cys2652Tyr)	FBN1	15	48415632	Marfan syndrome;not provided
NM_000138.4(FBN1):c.3037G>A (p.Gly1013Arg)	FBN1	15	48489896	Marfan syndrome;not provided
NM_000138.4(FBN1):c.8080C>T (p.Arg2694Ter)	FBN1	15	48412715	Marfan syndrome;not provided
NM_000138.4(FBN1):c.1633C>T (p.Arg545Cys)	FBN1	15	48510125	Marfan syndrome;not provided
NM_000138.4(FBN1):c.7205-1G>A	FBN1	15	48425865	Marfan syndrome;not provided
NM_000138.4(FBN1):c.4621C>T (p.Arg1541Ter)	FBN1	15	48468064	Marfan syndrome;not provided
NM_000138.4(FBN1):c.1090C>T (p.Arg364Ter)	FBN1	15	48520716	Marfan syndrome;not provided
NM_000138.4(FBN1):c.4781G>A (p.Gly1594Asp)	FBN1	15	48465825	Marfan syndrome;Stiff skin syndrome
NM_000138.4(FBN1):c.3668G>A (p.Cys1223Tyr)	FBN1	15	48485418	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.643C>T (p.Arg215Ter)	FBN1	15	48537704	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.8326C>T (p.Arg2776Ter)	FBN1	15	48411280	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.6354C>T (p.Ile2118=)	FBN1	15	48437347	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.1468+5G>A	FBN1	15	48515382	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.1546C>T (p.Arg516Ter)	FBN1	15	48513591	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.4615C>T (p.Arg1539Ter)	FBN1	15	48468070	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.5368C>T (p.Arg1790Ter)	FBN1	15	48456691	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.1285C>T (p.Arg429Ter)	FBN1	15	48516225	Marfan syndrome;Thoracic aortic aneurysm and aortic dissection;not provided
NM_022464.4(SIL1):c.645+1G>A	SIL1	5	139026800	Marinesco-SjÃ¶gren syndrome
NM_001037633.1(SIL1):c.331C>T (p.Arg111Ter)	SIL1	5	139050960	Marinesco-SjÃ¶gren syndrome
NM_022464.4(SIL1):c.865_1029del	SIL1	5	138951170	Marinesco-SjÃ¶gren syndrome
NM_001037633.1(SIL1):c.460C>T (p.Gln154Ter)	SIL1	5	139026986	Marinesco-SjÃ¶gren syndrome
NM_001037633.1(SIL1):c.1312C>T (p.Gln438Ter)	SIL1	5	138947191	Marinesco-SjÃ¶gren syndrome;not provided
NM_080629.2(COL11A1):c.3852+1G>A	COL11A1	1	102915630	Marshall syndrome
NM_002501.3(NFIX):c.955+1G>A	NFIX	19	13075672	Marshall-Smith syndrome
NM_012414.3(RAB3GAP2):c.1276C>T (p.Arg426Cys)	RAB3GAP2	1	220191279	Martsolf syndrome
NM_000457.4(HNF4A):c.829C>T (p.Gln277Ter)	HNF4A	20	44419813	Maturity-onset diabetes of the young, type 1
NM_000457.4(HNF4A):c.487C>T (p.Arg163Ter)	HNF4A	20	44413795	Maturity-onset diabetes of the young, type 1
NM_000457.4(HNF4A):c.406C>T (p.Arg136Trp)	HNF4A	20	44413714	Maturity-onset diabetes of the young, type 1
NM_000162.3(GCK):c.1132G>A (p.Ala378Thr)	GCK	7	44145618	Maturity-onset diabetes of the young, type 2
NM_000162.3(GCK):c.1015G>A (p.Glu339Lys)	GCK	7	44146467	Maturity-onset diabetes of the young, type 2
NM_000162.3(GCK):c.118G>A (p.Glu40Lys)	GCK	7	44153391	Maturity-onset diabetes of the young, type 2
NM_000162.3(GCK):c.112C>T (p.Gln38Ter)	GCK	7	44153397	Maturity-onset diabetes of the young, type 2
NM_000162.3(GCK):c.781G>A (p.Gly261Arg)	GCK	7	44147732	Maturity-onset diabetes of the young, type 2;not provided
NM_000162.3(GCK):c.214G>A (p.Gly72Arg)	GCK	7	44152420	Maturity-onset diabetes of the young, type 2;not provided
NM_000162.3(GCK):c.676G>A (p.Val226Met)	GCK	7	44149763	Maturity-onset diabetes of the young, type 2;not provided
NM_000162.3(GCK):c.76C>T (p.Gln26Ter)	GCK	7	44153433	Maturity-onset diabetes of the young, type 2;not provided
NM_000162.3(GCK):c.683C>T (p.Thr228Met)	GCK	7	44147830	Maturity-onset diabetes of the young, type 2;Permanent neonatal diabetes mellitus
NM_000207.2(INS):c.16C>T (p.Arg6Cys)	INS	11	2160956	Maturity-onset diabetes of the young, type 10
NM_000207.2(INS):c.137G>A (p.Arg46Gln)	INS	11	2160835	Maturity-onset diabetes of the young, type 10
NC_000008.11:g.11573132C>T	BLK	8	11573132	Maturity-onset diabetes of the young, type 11
NC_000008.11:g.11474238G>A	BLK	8	11474238	Maturity-onset diabetes of the young, type 11
NM_012096.2(APPL1):c.280G>A (p.Asp94Asn)	APPL1	3	57238111	Maturity-onset diabetes of the young, type 14
NM_000545.6(HNF1A):c.1340C>T (p.Pro447Leu)	HNF1A	12	120997504	Maturity-onset diabetes of the young, type 3
NM_000545.6(HNF1A):c.1859C>T (p.Thr620Ile)	HNF1A	12	121001155	Maturity-onset diabetes of the young, type 3

NM_000545.6(HNF1A):c.335C>T (p.Pro112Leu)	HNF1A	12	120988841	Maturity-onset diabetes of the young, type 3
NM_000545.6(HNF1A):c.391C>T (p.Arg131Trp)	HNF1A	12	120988897	Maturity-onset diabetes of the young, type 3;not provided
NM_006193.2(PAX4):c.490C>T (p.Arg164Trp)	PAX4	7	127613804	Maturity-onset diabetes of the young, type 9
NM_021083.3(XK):c.941G>A (p.Trp314Ter)	XK	X	37728068	McLeod neuroacanthocytosis syndrome
NM_021083.3(XK):c.895C>T (p.Gln299Ter)	XK	X	37728022	McLeod neuroacanthocytosis syndrome
NM_024426.4(WT1):c.1300C>T (p.Arg434Cys)	WT1	11	32392705	Meacham syndrome
NM_017777.3(MKS1):c.1024+1G>A	MKS1	17	58210658	Meckel syndrome type 1
NM_017777.3(MKS1):c.844C>T (p.Arg282Ter)	MKS1	17	58212996	Meckel syndrome type 1
NM_153704.5(TMEM67):c.1575+1G>A	TMEM67	8	93791320	Meckel syndrome type 3
NM_025114.3(CEP290):c.613C>T (p.Arg205Ter)	CEP290	12	88130324	Meckel syndrome type 4
NM_001127897.3(RPGRIP1L):c.1033C>T (p.Gln345Ter)	RPGRIP1L	16	53671580	Meckel syndrome type 5
NM_001077416.2(TMEM231):c.823G>A (p.Val275Ile)	TMEM231	16	75542602	Meckel syndrome, type 11;Meckel-Gruber syndrome
NM_001080522.2(CC2D2A):c.394C>T (p.Arg132Ter)	CC2D2A	4	15502879	Meckel-Gruber syndrome
NM_017777.3(MKS1):c.1066C>T (p.Gln356Ter)	MKS1	17	58208542	Meckel-Gruber syndrome;not provided
NM_004963.3(GUCY2C):c.2008G>A (p.Ala670Thr)	GUCY2C	12	14641142	Meconium ileus
NM_000016.5(ACADM):c.447G>A (p.Met149Ile)	ACADM	1	75734850	Medium-chain acyl-coenzyme A dehydrogenase deficiency
NM_000016.5(ACADM):c.347G>A (p.Cys116Tyr)	ACADM	1	75733588	Medium-chain acyl-coenzyme A dehydrogenase deficiency
NM_000016.5(ACADM):c.469-1G>A	ACADM	1	75739979	Medium-chain acyl-coenzyme A dehydrogenase deficiency
NM_000016.5(ACADM):c.1012C>T (p.Gln338Ter)	ACADM	1	75761188	Medium-chain acyl-coenzyme A dehydrogenase deficiency
NM_000016.5(ACADM):c.387+1G>A	ACADM	1	75733629	Medium-chain acyl-coenzyme A dehydrogenase deficiency
NM_000016.5(ACADM):c.799G>A (p.Gly267Arg)	ACADM	1	75749509	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_000016.5(ACADM):c.583G>A (p.Gly195Arg)	ACADM	1	75740094	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_000016.5(ACADM):c.734C>T (p.Ser245Leu)	ACADM	1	75749444	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_000016.5(ACADM):c.362C>T (p.Thr121Ile)	ACADM	1	75733603	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_000016.5(ACADM):c.157C>T (p.Arg53Cys)	ACADM	1	75732682	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_000016.5(ACADM):c.617G>A (p.Arg206His)	ACADM	1	75745823	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_000016.5(ACADM):c.1045C>T (p.Arg349Ter)	ACADM	1	75761221	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_000016.5(ACADM):c.250C>T (p.Leu84Phe)	ACADM	1	75732886	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_000016.5(ACADM):c.600-18G>A	ACADM	1	75745788	Medium-chain acyl-coenzyme A dehydrogenase deficiency;not provided
NM_020630.4(RET):c.1859G>A (p.Cys620Tyr)	RET	10	43113655	Medullary thyroid carcinoma;Multiple endocrine neoplasia, type 1;Multiple endocrine neoplasia, type 2a;Multiple endocrine neoplasia, type 2b;Multiple endocrine neoplasia, type 4;not provided
NM_016169.3(SUFU):c.44C>T (p.Pro15Leu)	SUFU	10	102504196	Medulloblastoma
NM_057088.2(KRT3):c.1525G>A (p.Glu509Lys)	KRT3	12	52791216	Meesman's corneal dystrophy;not provided
NM_001711.5(BGN):c.5G>A (p.Trp2Ter)	BGN	X	153504636	MEESTER-LOEYS SYNDROME;Thoracic aortic aneurysm and aortic dissection
NM_001711.5(BGN):c.238G>A (p.Gly80Ser)	BGN	X	153504869	MEESTER-LOEYS SYNDROME;Thoracic aortic aneurysm and aortic dissection
NM_015166.3(MLC1):c.839C>T (p.Ser280Leu)	MLC1	22	50068488	Megalencephalic leukoencephalopathy with subcortical cysts 1
NM_015166.3(MLC1):c.278C>T (p.Ser93Leu)	MLC1	22	50080387	Megalencephalic leukoencephalopathy with subcortical cysts 1
NM_015166.3(MLC1):c.176G>A (p.Gly59Glu)	MLC1	22	50084727	Megalencephalic leukoencephalopathy with subcortical cysts 1;not provided
NM_015166.3(MLC1):c.206C>T (p.Ser69Leu)	MLC1	22	50083145	Megalencephalic leukoencephalopathy with subcortical cysts 1;not provided
NM_152722.4(HEPACAM):c.275G>A (p.Arg92Gln)	HEPACAM	11	124924880	Megalencephalic leukoencephalopathy with subcortical cysts 2a
NM_152722.4(HEPACAM):c.631G>A (p.Asp211Asn)	HEPACAM	11	124923807	Megalencephalic leukoencephalopathy with subcortical cysts 2a
NM_152722.4(HEPACAM):c.292C>T (p.Arg98Cys)	HEPACAM	11	124924863	Megalencephalic leukoencephalopathy with subcortical cysts 2a
NM_152722.4(HEPACAM):c.274C>T (p.Arg92Trp)	HEPACAM	11	124924881	Megalencephalic leukoencephalopathy with subcortical cysts 2a;Megalencephalic leukoencephalopathy with subcortical cysts 2b, remitting, with or without mental retardation

NM_152722.4(HEPACAM):c.265G>A (p.Gly89Ser)	HEPACAM	11	124924890	Megalencephalic leukoencephalopathy with subcortical cysts 2a;Megalencephalic leukoencephalopathy with subcortical cysts 2b, remitting, with or without mental retardation;not provided
NM_152722.4(HEPACAM):c.266G>A (p.Gly89Asp)	HEPACAM	11	124924889	Megalencephalic leukoencephalopathy with subcortical cysts 2b, remitting, with mental retardation
NM_015166.3(MLC1):c.274C>T (p.Pro92Ser)	MLC1	22	50080391	Megalencephalic leukoencephalopathy with subcortical cysts;Megalencephalic leukoencephalopathy with subcortical cysts 1
NM_006218.3(PIK3CA):c.2740G>A (p.Gly914Arg)	PIK3CA	3	179230077	Megalencephaly cutis marmorata telangiectatica congenita;not provided
NM_006218.3(PIK3CA):c.1133G>A (p.Cys378Tyr)	PIK3CA	3	179204576	Megalencephaly cutis marmorata telangiectatica congenita;PIK3CA related overgrowth spectrum
NM_181690.2(AKT3):c.49G>A (p.Glu17Lys)	AKT3	1	243695714	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2
NM_001759.3(CCND2):c.841C>T (p.Pro281Ser)	CCND2	12	4299980	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3
NM_001759.3(CCND2):c.842C>T (p.Pro281Leu)	CCND2	12	4299981	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3
NM_001081.3(CUBN):c.3890C>T (p.Pro1297Leu)	CUBN	10	17041160	Megaloblastic anemia 1, Finnish type;Megaloblastic anemia due to inborn errors of metabolism
NM_030943.3(AMN):c.122C>T (p.Thr41Ile)	AMN	14	102923789	Megaloblastic anemia 1, Norwegian type
NM_000791.3(DHFR):c.238C>T (p.Leu80Phe)	DHFR	5	80649393	Megaloblastic anemia due to dihydrofolate reductase deficiency
NM_006996.2(SLC19A2):c.484C>T (p.Arg162Ter)	SLC19A2	1	169477478	Megaloblastic anemia, thiamine-responsive, with diabetes mellitus and sensorineural deafness
NM_006996.2(SLC19A2):c.750G>A (p.Trp250Ter)	SLC19A2	1	169477212	Megaloblastic anemia, thiamine-responsive, with diabetes mellitus and sensorineural deafness
NM_006996.2(SLC19A2):c.1074G>A (p.Trp358Ter)	SLC19A2	1	169468793	Megaloblastic anemia, thiamine-responsive, with diabetes mellitus and sensorineural deafness
NM_006996.2(SLC19A2):c.152C>T (p.Pro51Leu)	SLC19A2	1	169485615	Megaloblastic anemia, thiamine-responsive, with diabetes mellitus and sensorineural deafness
NM_006996.2(SLC19A2):c.515G>A (p.Gly172Asp)	SLC19A2	1	169477447	Megaloblastic anemia, thiamine-responsive, with diabetes mellitus and sensorineural deafness;not provided
NM_001143981.1(CHRDL1):c.652C>T (p.Arg218Ter)	CHRDL1	X	110694289	Megalocornea
NM_004153.3(ORC1):c.2159G>A (p.Arg720Gln)	ORC1	1	52375574	Meier-Gorlin syndrome 1
NM_004153.3(ORC1):c.1996C>T (p.Arg666Trp)	ORC1	1	52383437	Meier-Gorlin syndrome 1
NM_014321.3(ORC6):c.449+5G>A	ORC6	16	46693187	Meier-Gorlin syndrome 3
NM_030928.3(CDT1):c.196G>A (p.Ala66Thr)	CDT1	16	88804027	Meier-Gorlin syndrome 4
NM_030928.3(CDT1):c.1402G>A (p.Glu468Lys)	CDT1	16	88807407	Meier-Gorlin syndrome 4
NM_030928.3(CDT1):c.832+1G>A	CDT1	16	88805870	Meier-Gorlin syndrome 4
NM_030928.3(CDT1):c.1385G>A (p.Arg462Gln)	CDT1	16	88807390	Meier-Gorlin syndrome 4;not provided
NM_003504.4(CDC45):c.318C>T (p.Val106=)	CDC45	22	19482803	Meier-gorlin syndrome 7
NM_003504.4(CDC45):c.469C>T (p.Arg157Cys)	CDC45	22	19483988	Meier-gorlin syndrome 7
NM_003504.4(CDC45):c.1660C>T (p.Arg554Trp)	CDC45	22	19518867	Meier-gorlin syndrome 7
NM_003504.4(CDC45):c.333C>T (p.Asn111=)	CDC45	22	19482818	Meier-gorlin syndrome 7
NM_003504.4(CDC45):c.893C>T (p.Ala298Val)	CDC45	22	19507454	Meier-gorlin syndrome 7
NM_004153.3(ORC1):c.314G>A (p.Arg105Gln)	ORC1	1	52397773	Meier-Gorlin syndrome;Meier-Gorlin syndrome 1
NM_015450.2(POT1):c.1687-1G>A	POT1	7	124825358	Melanoma, cutaneous malignant, susceptibility to, 10
NM_015450.2(POT1):c.410G>A (p.Arg137His)	POT1	7	124863486	Melanoma, cutaneous malignant, susceptibility to, 10
NM_015450.2(POT1):c.809G>A (p.Ser270Asn)	POT1	7	124853032	Melanoma, cutaneous malignant, susceptibility to, 10
NM_015450.2(POT1):c.670G>A (p.Asp224Asn)	POT1	7	124858989	Melanoma, cutaneous malignant, susceptibility to, 10
NM_000503.5(EYA1):c.922C>T (p.Arg308Ter)	EYA1	8	71271802	Melnick-Fraser syndrome
NM_000503.5(EYA1):c.1319G>A (p.Arg440Gln)	EYA1	8	71216733	Melnick-Fraser syndrome
NM_172060.3(EYA1):c.867+5G>A	EYA1	8	71271753	Melnick-Fraser syndrome
NM_005982.3(SIX1):c.334C>T (p.Arg112Cys)	SIX1	14	60648856	Melnick-Fraser syndrome
NM_000503.5(EYA1):c.428G>A (p.Trp143Ter)	EYA1	8	71317680	Melnick-Fraser syndrome
NM_001110556.1(FLNA):c.3562G>A (p.Ala1188Thr)	FLNA	X	154360233	Melnick-Needles syndrome
NM_001110556.1(FLNA):c.3596C>T (p.Ser1199Leu)	FLNA	X	154360199	Melnick-Needles syndrome;Oto-palato-digital syndrome, type II

NM_020630.4(RET):c.2647G>A (p.Ala883Thr)	RET	10	43120120	MEN2 phenotype: Unclassified
NM_020630.4(RET):c.2711C>T (p.Ser904Phe)	RET	10	43120184	MEN2 phenotype: Unclassified
NM_016169.3(SUFU):c.367C>T (p.Arg123Cys)	SUFU	10	102550019	Meningioma, familial
NM_003079.4(SMARCE1):c.311G>A (p.Trp104Ter)	SMARCE1	17	40636453	Meningioma, familial
NM_003079.4(SMARCE1):c.715C>T (p.Arg239Ter)	SMARCE1	17	40631693	Meningioma, familial
NM_003079.4(SMARCE1):c.313C>T (p.Arg105Ter)	SMARCE1	17	40636451	Meningioma, familial
NM_000268.3(NF2):c.169C>T (p.Arg57Ter)	NF2	22	29636805	Meningioma;not provided
NM_000052.6(ATP7A):c.2938C>T (p.Arg980Ter)	ATP7A	X	78029271	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.3056G>A (p.Gly1019Asp)	ATP7A	X	78029389	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.601C>T (p.Arg201Ter)	ATP7A	X	77988722	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.1947-1G>A	ATP7A	X	78011448	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.598C>T (p.Gln200Ter)	ATP7A	X	77988719	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.1225C>T (p.Arg409Ter)	ATP7A	X	77989847	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.1544-1G>A	ATP7A	X	78003072	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.1639C>T (p.Arg547Ter)	ATP7A	X	78003168	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.1933C>T (p.Arg645Ter)	ATP7A	X	78011239	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.1946+5G>A	ATP7A	X	78011257	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.1950G>A (p.Trp650Ter)	ATP7A	X	78011452	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.2179G>A (p.Gly727Arg)	ATP7A	X	78012885	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.2187G>A (p.Trp729Ter)	ATP7A	X	78012893	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.2383C>T (p.Arg795Ter)	ATP7A	X	78013089	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.2499-1G>A	ATP7A	X	78015753	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.2555C>T (p.Pro852Leu)	ATP7A	X	78015810	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.2956C>T (p.Arg986Ter)	ATP7A	X	78029289	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.3112-1G>A	ATP7A	X	78031399	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.3466C>T (p.Gln1156Ter)	ATP7A	X	78033776	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.3502C>T (p.Gln1168Ter)	ATP7A	X	78033812	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.3764G>A (p.Gly1255Glu)	ATP7A	X	78040696	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.3943G>A (p.Gly1315Arg)	ATP7A	X	78042726	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.4123+1G>A	ATP7A	X	78043435	Menkes kinky-hair syndrome
NM_000052.6(ATP7A):c.4226+5G>A	ATP7A	X	78045577	Menkes kinky-hair syndrome
NM_014271.3(IL1RAPL1):c.1460G>A (p.Trp487Ter)	IL1RAPL1	X	29955189	Mental retardation 21, X-linked
NM_005334.2(HCFC1):c.344C>T (p.Ala115Val)	HCFC1	X	153964283	Mental retardation 3, X-linked
NM_005334.2(HCFC1):c.217G>A (p.Ala73Thr)	HCFC1	X	153964703	Mental retardation 3, X-linked
NM_005334.2(HCFC1):c.674G>A (p.Ser225Asn)	HCFC1	X	153963263	Mental retardation 3, X-linked;not provided
NM_002578.4(PAK3):c.1255C>T (p.Arg419Ter)	PAK3	X	111196488	Mental retardation 30, X-linked
NM_002578.4(PAK3):c.199C>T (p.Arg67Cys)	PAK3	X	111142119	Mental retardation 30, X-linked
NM_001830.3(CLCN4):c.1630G>A (p.Gly544Arg)	CLCN4	X	10213734	Mental retardation 49, X-linked
NM_022977.2(ACSL4):c.1124C>T (p.Pro375Leu)	ACSL4	X	109674403	Mental retardation 63, X-linked
NM_015335.4(MED13L):c.6485C>T (p.Thr2162Met)	MED13L	12	115963422	Mental retardation and distinctive facial features with or without cardiac defects
NM_003688.3(CASK):c.915G>A (p.Lys305=)	CASK	X	41636578	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
NM_003688.3(CASK):c.316C>T (p.Arg106Ter)	CASK	X	41745564	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
NM_003688.3(CASK):c.1639C>T (p.Gln547Ter)	CASK	X	41561588	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
NM_003688.3(CASK):c.2074C>T (p.Gln692Ter)	CASK	X	41542772	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
NM_003688.3(CASK):c.2470C>T (p.Gln824Ter)	CASK	X	41531042	Mental retardation and microcephaly with pontine and cerebellar hypoplasia

NM_003688.3(CASK):c.708+1G>A	CASK	X	41665276	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
NM_003688.3(CASK):c.880C>T (p.Gln294Ter)	CASK	X	41636613	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
NM_003688.3(CASK):c.1976G>A (p.Gly659Asp)	CASK	X	41553782	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
NM_003688.3(CASK):c.2377C>T (p.Gln793Ter)	CASK	X	41531135	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
NM_003688.3(CASK):c.1915C>T (p.Arg639Ter)	CASK	X	41553843	Mental retardation and microcephaly with pontine and cerebellar hypoplasia;not provided
NM_003688.3(CASK):c.2041C>T (p.Arg681Ter)	CASK	X	41542805	Mental retardation and microcephaly with pontine and cerebellar hypoplasia;not provided
NM_003688.3(CASK):c.82C>T (p.Arg28Ter)	CASK	X	41853205	Mental retardation and microcephaly with pontine and cerebellar hypoplasia;not provided
NM_032682.5(FOXP1):c.1573C>T (p.Arg525Ter)	FOXP1	3	70972634	Mental retardation with language impairment and with or without autistic features
NM_032682.5(FOXP1):c.1624C>T (p.Gln542Ter)	FOXP1	3	70972583	Mental retardation with language impairment and with or without autistic features
NM_032682.5(FOXP1):c.1540C>T (p.Arg514Cys)	FOXP1	3	70972667	Mental retardation with language impairment and with or without autistic features
NM_032682.5(FOXP1):c.1541G>A (p.Arg514His)	FOXP1	3	70972666	Mental retardation with language impairment and with or without autistic features;not provided
NM_032682.5(FOXP1):c.1507C>T (p.Arg503Ter)	FOXP1	3	70976964	Mental retardation with language impairment and with or without autistic features;not provided
NM_002547.2(OPHN1):c.184C>T (p.Gln62Ter)	OPHN1	X	68299067	Mental retardation X-linked with cerebellar hypoplasia and distinctive facial appearance
NM_002547.2(OPHN1):c.496C>T (p.Gln166Ter)	OPHN1	X	68213963	Mental retardation X-linked with cerebellar hypoplasia and distinctive facial appearance
NM_018013.3(SOBP):c.1981C>T (p.Arg661Ter)	SOBP	6	107634825	Mental retardation, anterior maxillary protrusion, and strabismus
NM_020732.3(ARID1B):c.2149C>T (p.Gln717Ter)	ARID1B	6	157084773	Mental retardation, autosomal dominant 12
NM_020732.3(ARID1B):c.2248C>T (p.Arg750Ter)	ARID1B	6	157084872	Mental retardation, autosomal dominant 12
NM_020732.3(ARID1B):c.5404C>T (p.Arg1802Ter)	ARID1B	6	157206545	Mental retardation, autosomal dominant 12
NM_020732.3(ARID1B):c.5968C>T (p.Arg1990Ter)	ARID1B	6	157207109	Mental retardation, autosomal dominant 12
NM_020732.3(ARID1B):c.6100C>T (p.Gln2034Ter)	ARID1B	6	157207241	Mental retardation, autosomal dominant 12
NM_020732.3(ARID1B):c.2242C>T (p.Gln748Ter)	ARID1B	6	157084866	Mental retardation, autosomal dominant 12
NM_020732.3(ARID1B):c.2692C>T (p.Arg898Ter)	ARID1B	6	157148764	Mental retardation, autosomal dominant 12;not provided
NM_001376.4(DYNC1H1):c.4552G>A (p.Glu1518Lys)	DYNC1H1	14	102002546	Mental retardation, autosomal dominant 13
NM_001376.4(DYNC1H1):c.10151G>A (p.Arg3384Gln)	DYNC1H1	14	102033136	Mental retardation, autosomal dominant 13
NM_001376.4(DYNC1H1):c.10031G>A (p.Arg3344Gln)	DYNC1H1	14	102032419	Mental retardation, autosomal dominant 13
NM_001376.4(DYNC1H1):c.10573C>T (p.Arg3525Cys)	DYNC1H1	14	102034135	Mental retardation, autosomal dominant 13
NM_001376.4(DYNC1H1):c.926G>A (p.Arg309His)	DYNC1H1	14	101980515	Mental retardation, autosomal dominant 13;not provided
NM_006015.4(ARID1A):c.2758C>T (p.Gln920Ter)	ARID1A	1	26766246	Mental retardation, autosomal dominant 14
NM_006015.4(ARID1A):c.4003C>T (p.Arg1335Ter)	ARID1A	1	26773716	Mental retardation, autosomal dominant 14
NM_006015.4(ARID1A):c.5965C>T (p.Arg1989Ter)	ARID1A	1	26779863	Mental retardation, autosomal dominant 14
NM_003073.4(SMARCB1):c.1130G>A (p.Arg377His)	SMARCB1	22	23834152	Mental retardation, autosomal dominant 15
NM_003073.4(SMARCB1):c.110G>A (p.Arg37His)	SMARCB1	22	23791772	Mental retardation, autosomal dominant 15;not provided
NM_001128849.1(SMARCA4):c.2851G>A (p.Gly951Arg)	SMARCA4	19	11021959	Mental retardation, autosomal dominant 16
NM_003072.3(SMARCA4):c.2576C>T (p.Thr859Met)	SMARCA4	19	11019661	Mental retardation, autosomal dominant 16;not provided
NM_003072.3(SMARCA4):c.2653C>T (p.Arg885Cys)	SMARCA4	19	11021761	Mental retardation, autosomal dominant 16;not provided
NM_003072.3(SMARCA4):c.2761C>T (p.Leu921Phe)	SMARCA4	19	11021869	Mental retardation, autosomal dominant 16;not provided
NM_020699.3(GATAD2B):c.1408C>T (p.Gln470Ter)	GATAD2B	1	153813261	Mental retardation, autosomal dominant 18
NM_020699.3(GATAD2B):c.694C>T (p.Gln232Ter)	GATAD2B	1	153818075	Mental retardation, autosomal dominant 18
NM_001904.3(CTNNB1):c.925C>T (p.Gln309Ter)	CTNNB1	3	41225850	Mental retardation, autosomal dominant 19
NM_001904.3(CTNNB1):c.268C>T (p.Arg90Ter)	CTNNB1	3	41224980	Mental retardation, autosomal dominant 19
NM_001904.3(CTNNB1):c.1543C>T (p.Arg515Ter)	CTNNB1	3	41234157	Mental retardation, autosomal dominant 19;not provided
NM_001904.3(CTNNB1):c.283C>T (p.Arg95Ter)	CTNNB1	3	41224995	Mental retardation, autosomal dominant 19;not provided
NM_006565.3(CTCF):c.1699C>T (p.Arg567Trp)	CTCF	16	67628550	Mental retardation, autosomal dominant 21;not provided
NM_001080517.2(SETD5):c.3001C>T (p.Arg1001Ter)	SETD5	3	9470735	Mental retardation, autosomal dominant 23
NM_001080517.2(SETD5):c.2302C>T (p.Arg768Ter)	SETD5	3	9448586	Mental retardation, autosomal dominant 23

NM_021008.3(DEAF1):c.670C>T (p.Arg224Trp)	DEAF1	11	686992	Mental retardation, autosomal dominant 24
NM_021008.3(DEAF1):c.634G>A (p.Gly212Ser)	DEAF1	11	687941	Mental retardation, autosomal dominant 24
NM_003108.3(SOX11):c.1286G>A (p.Trp429Ter)	SOX11	2	5694007	Mental retardation, autosomal dominant 27
NM_015559.2(SETBP1):c.1596G>A (p.Trp532Ter)	SETBP1	18	44950936	Mental retardation, autosomal dominant 29
NM_015559.2(SETBP1):c.1873C>T (p.Arg625Ter)	SETBP1	18	44951213	Mental retardation, autosomal dominant 29;not provided
NM_015559.2(SETBP1):c.1876C>T (p.Arg626Ter)	SETBP1	18	44951216	Mental retardation, autosomal dominant 29;not provided
NM_004933.2(CDH15):c.178C>T (p.Arg60Cys)	CDH15	16	89179551	Mental retardation, autosomal dominant 3
NM_004933.2(CDH15):c.274C>T (p.Arg92Trp)	CDH15	16	89180272	Mental retardation, autosomal dominant 3
NM_004933.2(CDH15):c.365C>T (p.Ala122Val)	CDH15	16	89183555	Mental retardation, autosomal dominant 3
NM_006624.5(ZMYND11):c.976C>T (p.Gln326Ter)	ZMYND11	10	246791	Mental retardation, autosomal dominant 30
NM_001202468.1(ZMYND11):c.1262G>A (p.Ser421Asn)	ZMYND11	10	248370	Mental retardation, autosomal dominant 30
NM_006624.5(ZMYND11):c.22C>T (p.Arg8Ter)	ZMYND11	10	180034	Mental retardation, autosomal dominant 30
NM_005859.4(PURA):c.367C>T (p.Gln123Ter)	PURA	5	140114548	Mental retardation, autosomal dominant 31
NM_006766.4(KAT6A):c.3505C>T (p.Arg1169Ter)	KAT6A	8	41934715	Mental retardation, autosomal dominant 32;not provided
NM_006245.3(PPP2R5D):c.157C>T (p.Pro53Ser)	PPP2R5D	6	43006514	Mental retardation, autosomal dominant 35
NM_006245.3(PPP2R5D):c.592G>A (p.Glu198Lys)	PPP2R5D	6	43007265	Mental retardation, autosomal dominant 35;not provided
NM_006245.3(PPP2R5D):c.598G>A (p.Glu200Lys)	PPP2R5D	6	43007271	Mental retardation, autosomal dominant 35;not provided
NM_014225.5(PPP2R1A):c.544C>T (p.Arg182Trp)	PPP2R1A	19	52212726	Mental retardation, autosomal dominant 36;not provided
NM_014225.5(PPP2R1A):c.773G>A (p.Arg258His)	PPP2R1A	19	52213076	Mental retardation, autosomal dominant 36;not provided
NM_015025.3(MYT1L):c.2636+1G>A	MYT1L	2	1887487	Mental retardation, autosomal dominant 39;not provided
NM_032531.3(KIRREL3):c.118C>T (p.Arg40Trp)	KIRREL3	11	126562850	Mental retardation, autosomal dominant 4
NM_032531.3(KIRREL3):c.1007G>A (p.Arg336Gln)	KIRREL3	11	126446877	Mental retardation, autosomal dominant 4
NM_032436.3(CHAMP1):c.1002G>A (p.Trp334Ter)	CHAMP1	13	114324844	Mental retardation, autosomal dominant 40
NM_032436.3(CHAMP1):c.1489C>T (p.Arg497Ter)	CHAMP1	13	114325331	Mental retardation, autosomal dominant 40
NM_032436.3(CHAMP1):c.1192C>T (p.Arg398Ter)	CHAMP1	13	114325034	Mental retardation, autosomal dominant 40;intellectual disability with severe speech impairment
NM_032436.3(CHAMP1):c.1768C>T (p.Gln590Ter)	CHAMP1	13	114325610	Mental retardation, autosomal dominant 40;intellectual disability with severe speech impairment
NM_006734.3(HIVEP2):c.2827C>T (p.Arg943Ter)	HIVEP2	6	142771912	Mental retardation, autosomal dominant 43
NM_006734.3(HIVEP2):c.3556C>T (p.Gln1186Ter)	HIVEP2	6	142771183	Mental retardation, autosomal dominant 43
NM_006772.2(SYNGAP1):c.427C>T (p.Arg143Ter)	SYNGAP1	6	33432724	Mental retardation, autosomal dominant 5
NM_006772.2(SYNGAP1):c.1861C>T (p.Arg621Ter)	SYNGAP1	6	33440913	Mental retardation, autosomal dominant 5
NM_006772.2(SYNGAP1):c.2059C>T (p.Arg687Ter)	SYNGAP1	6	33441318	Mental retardation, autosomal dominant 5
NM_006772.2(SYNGAP1):c.1735C>T (p.Arg579Ter)	SYNGAP1	6	33440787	Mental retardation, autosomal dominant 5;not provided
NM_006772.2(SYNGAP1):c.1685C>T (p.Pro562Leu)	SYNGAP1	6	33440737	Mental retardation, autosomal dominant 5;not provided
NM_000834.3(GRIN2B):c.411+1G>A	GRIN2B	12	13865797	Mental retardation, autosomal dominant 6
NM_000834.3(GRIN2B):c.2044C>T (p.Arg682Cys)	GRIN2B	12	13571931	Mental retardation, autosomal dominant 6
NM_000834.3(GRIN2B):c.1367G>A (p.Cys456Tyr)	GRIN2B	12	13615626	Mental retardation, autosomal dominant 6
NM_000834.3(GRIN2B):c.1677G>A (p.Trp559Ter)	GRIN2B	12	13611828	Mental retardation, autosomal dominant 6
NM_000834.3(GRIN2B):c.1658C>T (p.Pro553Leu)	GRIN2B	12	13611847	Mental retardation, autosomal dominant 6
NM_000834.3(GRIN2B):c.2065G>A (p.Gly689Ser)	GRIN2B	12	13571910	Mental retardation, autosomal dominant 6;not provided
NM_001396.3(DYRK1A):c.1098+1G>A	DYRK1A	21	37493164	Mental retardation, autosomal dominant 7
NM_001396.3(DYRK1A):c.1399C>T (p.Arg467Ter)	DYRK1A	21	37505442	Mental retardation, autosomal dominant 7
NM_001396.3(DYRK1A):c.883C>T (p.Leu295Phe)	DYRK1A	21	37490393	Mental retardation, autosomal dominant 7;not provided
NM_001396.3(DYRK1A):c.1400G>A (p.Arg467Gln)	DYRK1A	21	37505443	Mental retardation, autosomal dominant 7;not provided
NM_001396.3(DYRK1A):c.1639C>T (p.Gln547Ter)	DYRK1A	21	37506191	Mental retardation, autosomal dominant 7;not provided
NM_007327.3(GRIN1):c.1984G>A (p.Glu662Lys)	GRIN1	9	137162710	Mental retardation, autosomal dominant 8

NM_007327.3(GRIN1):c.1923G>A (p.Met641Ile)	GRIN1	9	137162649	Mental retardation, autosomal dominant 8
NM_001244008.1(KIF1A):c.757G>A (p.Glu253Lys)	KIF1A	2	240783780	Mental retardation, autosomal dominant 9
NM_001244008.1(KIF1A):c.946C>T (p.Arg316Trp)	KIF1A	2	240775863	Mental retardation, autosomal dominant 9;not provided
NM_001244008.1(KIF1A):c.647G>A (p.Arg216His)	KIF1A	2	240785062	Mental retardation, autosomal dominant 9;PEHO syndrome
NM_001244008.1(KIF1A):c.646C>T (p.Arg216Cys)	KIF1A	2	240785063	Mental retardation, autosomal dominant 9;PEHO syndrome
NM_001244008.1(KIF1A):c.296C>T (p.Thr99Met)	KIF1A	2	240788118	Mental retardation, autosomal dominant 9;PEHO syndrome;not provided
NM_001160372.3(TRAPPC9):c.1129C>T (p.Arg377Ter)	TRAPPC9	8	140397625	Mental retardation, autosomal recessive 13
NM_031466.7(TRAPPC9):c.1708C>T (p.Arg570Ter)	TRAPPC9	8	140360131	Mental retardation, autosomal recessive 13;not provided
NM_138501.5(TECR):c.545C>T (p.Pro182Leu)	TECR	19	14564841	Mental retardation, autosomal recessive 14
NM_016219.4(MAN1B1):c.1189G>A (p.Glu397Lys)	MAN1B1	9	137101607	Mental retardation, autosomal recessive 15
NM_016219.4(MAN1B1):c.1000C>T (p.Arg334Cys)	MAN1B1	9	137101088	Mental retardation, autosomal recessive 15
NM_004830.3(MED23):c.3988C>T (p.Arg1330Ter)	MED23	6	131587798	Mental retardation, autosomal recessive 18
NM_015979.3(MED23):c.1850G>A (p.Arg617Gln)	MED23	6	131603129	Mental retardation, autosomal recessive 18;not provided
NM_016302.3(CRBN):c.1255C>T (p.Arg419Ter)	CRBN	3	3150939	Mental retardation, autosomal recessive 2
NM_003805.4(CRADD):c.508C>T (p.Arg170Cys)	CRADD	12	93850179	Mental retardation, autosomal recessive 34
NM_003805.4(CRADD):c.509G>A (p.Arg170His)	CRADD	12	93850180	Mental retardation, autosomal recessive 34
NM_138422.3(ADAT3):c.430G>A (p.Val144Met)	ADAT3	19	1912477	Mental retardation, autosomal recessive 36;not provided
NM_004667.5(HERC2):c.1781C>T (p.Pro594Leu)	HERC2	15	28265707	Mental retardation, autosomal recessive 38
NM_025115.3(TTI2):c.1100C>T (p.Pro367Leu)	TTI2	8	33503763	Mental retardation, autosomal recessive 39
NM_024989.3(PGAP1):c.1396C>T (p.Gln466Ter)	PGAP1	2	196875776	Mental retardation, autosomal recessive 42
NM_001080510.4(METTL23):c.397C>T (p.Gln133Ter)	METTL23	17	76733367	Mental retardation, autosomal recessive 44
NM_001543.4(NDST1):c.2126G>A (p.Arg709Gln)	NDST1	5	150545467	Mental retardation, autosomal recessive 46
NM_001543.4(NDST1):c.1831G>A (p.Gly611Ser)	NDST1	5	150541651	Mental retardation, autosomal recessive 46
NM_001010898.3(SLC6A17):c.484G>A (p.Gly162Arg)	SLC6A17	1	110174012	Mental retardation, autosomal recessive 48
NM_133443.3(GPT2):c.1210C>T (p.Arg404Ter)	GPT2	16	46922414	Mental retardation, autosomal recessive 49
NM_133443.3(GPT2):c.815C>T (p.Pro272Leu)	GPT2	16	46909922	Mental retardation, autosomal recessive 49
NM_017755.5(NSUN2):c.679C>T (p.Gln227Ter)	NSUN2	5	6620242	Mental retardation, autosomal recessive 5
NM_017755.5(NSUN2):c.1114C>T (p.Gln372Ter)	NSUN2	5	6611067	Mental retardation, autosomal recessive 5
NM_017755.5(NSUN2):c.2035G>A (p.Gly679Arg)	NSUN2	5	6600195	Mental retardation, autosomal recessive 5
NM_006895.2(HNMT):c.179G>A (p.Gly60Asp)	HNMT	2	137970206	Mental retardation, autosomal recessive 51
NM_030805.3(LMAN2L):c.158G>A (p.Arg53Gln)	LMAN2L	2	96739883	Mental retardation, autosomal recessive 52
NM_001127178.2(PIGG):c.928C>T (p.Gln310Ter)	PIGG	4	515999	Mental retardation, autosomal recessive 53
NM_001127178.2(PIGG):c.2005C>T (p.Arg669Cys)	PIGG	4	523849	Mental retardation, autosomal recessive 53
NM_001161563.2(TNIK):c.538C>T (p.Arg180Ter)	TNIK	3	171188803	Mental retardation, autosomal recessive 54
NM_031307.3(PUS3):c.1303C>T (p.Arg435Ter)	PUS3	11	125893928	Mental retardation, autosomal recessive 55
NM_024824.4(ZC3H14):c.460C>T (p.Arg154Ter)	ZC3H14	14	85572606	MENTAL RETARDATION, AUTOSOMAL RECESSIVE 56
NM_021956.4(GRIK2):c.592C>T (p.Arg198Ter)	GRIK2	6	101676673	Mental retardation, autosomal recessive 6
NM_006765.3(TUSC3):c.163C>T (p.Gln55Ter)	TUSC3	8	15623104	Mental retardation, autosomal recessive 7
NM_002397.4(MEF2C):c.565C>T (p.Arg189Ter)	MEF2C	5	88751881	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations
NM_001131005.2(MEF2C):c.71G>A (p.Arg24Lys)	MEF2C	5	88804785	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations
NM_080632.2(UPF3B):c.1288C>T (p.Arg430Ter)	UPF3B	X	119837771	Mental retardation, syndromic 14, X-linked
NM_004187.3(KDM5C):c.2191C>T (p.Leu731Phe)	KDM5C	X	53199029	Mental retardation, syndromic, Claes-Jensen type, X-linked
NM_004187.3(KDM5C):c.2080C>T (p.Arg694Ter)	KDM5C	X	53199140	Mental retardation, syndromic, Claes-Jensen type, X-linked
NM_004187.3(KDM5C):c.2296C>T (p.Arg766Trp)	KDM5C	X	53198836	Mental retardation, syndromic, Claes-Jensen type, X-linked
NM_004187.3(KDM5C):c.229G>A (p.Ala77Thr)	KDM5C	X	53218398	Mental retardation, syndromic, Claes-Jensen type, X-linked

NM_004187.3(KDM5C):c.3118C>T (p.Gln1040Ter)	KDM5C	X	53195918	Mental retardation, syndromic, Claes-Jensen type, X-linked	
NM_139058.2(ARX):c.856G>A (p.Gly286Ser)	ARX	X	25013139	Mental retardation, with or without seizures, ARX-related, X-linked	
NM_001111125.2(IQSEC2):c.2273G>A (p.Arg758Gln)	IQSEC2	X	53250303	Mental retardation, X-linked 1	
NM_001111125.2(IQSEC2):c.1075C>T (p.Arg359Cys)	IQSEC2	X	53254856	Mental retardation, X-linked 1	
NM_001111125.2(IQSEC2):c.2563C>T (p.Arg855Ter)	IQSEC2	X	53248133	Mental retardation, X-linked 1	
NM_001111125.2(IQSEC2):c.2366C>T (p.Ala789Val)	IQSEC2	X	53248814	Mental retardation, X-linked 1	
NM_012216.3(MID2):c.1040G>A (p.Arg347Gln)	MID2	X	107905593	Mental retardation, X-linked 101	
NM_001356.4(DDX3X):c.1126C>T (p.Arg376Cys)	DDX3X	X	41345280	Mental retardation, X-linked 102	
NM_001356.4(DDX3X):c.977G>A (p.Arg326His)	DDX3X	X	41344351	Mental retardation, X-linked 102	
NM_001356.4(DDX3X):c.1084C>T (p.Arg362Cys)	DDX3X	X	41345238	Mental retardation, X-linked 102	
NM_001356.4(DDX3X):c.619C>T (p.Gln207Ter)	DDX3X	X	41343291	Mental retardation, X-linked 102	
NM_001356.4(DDX3X):c.1052G>A (p.Arg351Gln)	DDX3X	X	41345206	Mental retardation, X-linked 102	
NM_001356.4(DDX3X):c.1703C>T (p.Pro568Leu)	DDX3X	X	41346946	Mental retardation, X-linked 102	
NM_001081550.1(THOC2):c.937C>T (p.Leu313Phe)	THOC2	X	123668239	Mental retardation, X-linked 12	
NM_004586.2(RPS6KA3):c.1147C>T (p.Arg383Trp)	RPS6KA3	X	20175244	Mental retardation, X-linked 19	
NM_171998.3(RAB39B):c.215+1G>A	RAB39B	X	155264073	Mental retardation, X-linked 72	
NM_153252.4(BRWD3):c.568C>T (p.Arg190Ter)	BRWD3	X	80745592	Mental retardation, X-linked 93	
NM_001039590.2(USP9X):c.2554C>T (p.Arg852Ter)	USP9X	X	41168136	Mental retardation, X-linked 99, syndromic, female-restricted	
NM_001039590.2(USP9X):c.3763C>T (p.Gln1255Ter)	USP9X	X	41188070	Mental retardation, X-linked 99, syndromic, female-restricted	
NM_001039590.2(USP9X):c.1111C>T (p.Arg371Ter)	USP9X	X	41141381	Mental retardation, X-linked 99, syndromic, female-restricted	
NM_004992.3(MECP2):c.674C>T (p.Pro225Leu)	MECP2	X	154031154	Mental retardation, X-linked, syndromic 13;Rett syndrome	
NM_004992.3(MECP2):c.964C>T (p.Pro322Ser)	MECP2	X	154030864	Mental retardation, X-linked, syndromic 13;Rett syndrome	
NM_004992.3(MECP2):c.763C>T (p.Arg255Ter)	MECP2	X	154031065	Mental retardation, X-linked, syndromic 13;Rett syndrome;not provided	
NM_004992.3(MECP2):c.730C>T (p.Gln244Ter)	MECP2	X	154031098	Mental retardation, X-linked, syndromic 13;Rett syndrome;not provided	
NM_004992.3(MECP2):c.1216C>T (p.Gln406Ter)	MECP2	X	154030612	Mental retardation, X-linked, syndromic 13;Rett syndrome;not provided	
NM_004606.4(TAF1):c.3736C>T (p.Arg1246Trp)	TAF1	X	71398627	Mental retardation, X-linked, syndromic 33	
NM_004606.4(TAF1):c.1786C>T (p.Pro596Ser)	TAF1	X	71382821	Mental retardation, X-linked, syndromic 33	
NM_001145408.1(NONO):c.1093C>T (p.Arg365Ter)	NONO	X	71297900	Mental retardation, X-linked, syndromic 34	
NM_001145408.1(NONO):c.1131G>A (p.Ala377=)	NONO	X	71297938	Mental retardation, X-linked, syndromic 34;not provided	
NM_001032393.2(HNRNPH2):c.616C>T (p.Arg206Trp)	HNRNPH2	X	101412604	MENTAL RETARDATION, X-LINKED, SYNDROMIC, BAIN TYPE	
NM_001032393.2(HNRNPH2):c.617G>A (p.Arg206Gln)	HNRNPH2	X	101412605	MENTAL RETARDATION, X-LINKED, SYNDROMIC, BAIN TYPE	
NM_005765.2(ATP6AP2):c.321C>T (p.Asp107=)	ATP6AP2	X	40597269	Mental retardation, X-linked, syndromic, Hedera type	
NM_001008222.2(ZDHHC9):c.442C>T (p.Arg148Trp)	ZDHHC9	X	129823724	Mental retardation, X-linked, syndromic, raymond type	
NM_001008222.2(ZDHHC9):c.448C>T (p.Pro150Ser)	ZDHHC9	X	129823718	Mental retardation, X-linked, syndromic, raymond type	
NM_031407.6(HUWE1):c.12037C>T (p.Arg4013Trp)	HUWE1	X	53537656	Mental retardation, X-linked, syndromic, turner type	
NM_031407.6(HUWE1):c.12559C>T (p.Arg4187Cys)	HUWE1	X	53535474	Mental retardation, X-linked, syndromic, turner type	
NM_031407.6(HUWE1):c.8942G>A (p.Arg2981His)	HUWE1	X	53551420	Mental retardation, X-linked, syndromic, turner type;not provided	
NM_007325.4(GRIA3):c.2497G>A (p.Gly833Arg)	GRIA3	X	123482856	Mental retardation, X-linked, syndromic, wu type	
NM_000489.4(ATRX):c.5131C>T (p.Pro1711Ser)	ATRX	X	77633210	Mental retardation-hypotonic facies syndrome, X-linked	
NM_000489.4(ATRX):c.659G>A (p.Cys220Tyr)	ATRX	X	77684942	Mental retardation-hypotonic facies syndrome, X-linked	
NM_007118.3(TRIO):c.4128G>A (p.Trp1376Ter)	TRIO	5	14390300	Mercer-Ba syndrome	
NM_007118.3(TRIO):c.4283G>A (p.Arg1428Gln)	TRIO	5	14394102	Mercer-Ba syndrome	
NM_001127666.1(GSN):c.520G>A (p.Asp174Asn)	GSN	9	121310819	Meretoja syndrome	
m.15967G>A	MT-TP	MT	15967	MERFF syndrome;Myoclonus with epilepsy with ragged red fibers	
NM_000426.3(LAMA2):c.9253C>T (p.Arg3085Ter)	LAMA2	6	129516231	Merosin deficient congenital muscular dystrophy	

NM_000426.3(LAMA2):c.7810C>T (p.Arg2604Ter)	LAMA2	6	129486534	Merosin deficient congenital muscular dystrophy
NM_000426.3(LAMA2):c.4048C>T (p.Arg1350Ter)	LAMA2	6	129316161	Merosin deficient congenital muscular dystrophy
NM_000426.3(LAMA2):c.7439+1G>A	LAMA2	6	129473353	Merosin deficient congenital muscular dystrophy
NM_000426.3(LAMA2):c.7155+1G>A	LAMA2	6	129464453	Merosin deficient congenital muscular dystrophy
NM_000426.3(LAMA2):c.5116C>T (p.Arg1706Ter)	LAMA2	6	129391535	Merosin deficient congenital muscular dystrophy
NM_000426.3(LAMA2):c.3718C>T (p.Gln1240Ter)	LAMA2	6	129315638	Merosin deficient congenital muscular dystrophy;not provided
NM_000426.3(LAMA2):c.7732C>T (p.Arg2578Ter)	LAMA2	6	129481422	Merosin deficient congenital muscular dystrophy;not provided
NM_000426.3(LAMA2):c.112+1G>A	LAMA2	6	128883358	Merosin deficient congenital muscular dystrophy;not provided
NM_000426.3(LAMA2):c.2962C>T (p.Gln988Ter)	LAMA2	6	129297790	Merosin deficient congenital muscular dystrophy;not provided
NM_000426.3(LAMA2):c.3976C>T (p.Arg1326Ter)	LAMA2	6	129316089	Merosin deficient congenital muscular dystrophy;not provided
NM_000426.3(LAMA2):c.4523+1G>A	LAMA2	6	129349385	Merosin deficient congenital muscular dystrophy;not provided
NM_000426.3(LAMA2):c.5914C>T (p.Gln1972Ter)	LAMA2	6	129427800	Merosin deficient congenital muscular dystrophy;not provided
NM_000426.3(LAMA2):c.6955C>T (p.Arg2319Ter)	LAMA2	6	129460287	Merosin deficient congenital muscular dystrophy;not provided
NM_000426.3(LAMA2):c.9212-1G>A	LAMA2	6	129516189	Merosin deficient congenital muscular dystrophy;not provided
m.12207G>A	MT-TS2	MT	12207	MERRF/MELAS overlap syndrome
m.12147G>A	MT-TH	MT	12147	MERRF/MELAS overlap syndrome
NM_003868.2(FGF16):c.535C>T (p.Arg179Ter)	FGF16	X	77456433	Metacarpal 4-5 fusion
NM_002834.4(PTPN11):c.412C>T (p.Arg138Ter)	PTPN11	12	112453274	Metachondromatosis
NM_002834.4(PTPN11):c.1516C>T (p.Gln506Ter)	PTPN11	12	112489092	Metachondromatosis
NM_000487.5(ARSA):c.467G>A (p.Gly156Asp)	ARSA	22	50627051	Metachromatic leukodystrophy
NM_000487.5(ARSA):c.641C>T (p.Ala214Val)	ARSA	22	50626877	Metachromatic leukodystrophy
NM_000487.5(ARSA):c.736C>T (p.Arg246Cys)	ARSA	22	50626709	Metachromatic leukodystrophy
NM_000487.5(ARSA):c.868C>T (p.Arg290Cys)	ARSA	22	50626265	Metachromatic leukodystrophy
NM_000487.5(ARSA):c.986C>T (p.Thr329Ile)	ARSA	22	50626057	Metachromatic leukodystrophy
NM_000487.5(ARSA):c.346C>T (p.Arg116Ter)	ARSA	22	50627285	Metachromatic leukodystrophy
NM_000487.5(ARSA):c.1229C>T (p.Thr410Ile)	ARSA	22	50625446	Metachromatic leukodystrophy, adult type
NM_000487.5(ARSA):c.931G>A (p.Gly311Ser)	ARSA	22	50626202	Metachromatic leukodystrophy, late infantile
NM_000487.5(ARSA):c.1115G>A (p.Arg372Gln)	ARSA	22	50625674	Metachromatic leukodystrophy, mild
NM_000487.5(ARSA):c.263G>A (p.Gly88Asp)	ARSA	22	50627368	Metachromatic leukodystrophy, severe
NM_000487.5(ARSA):c.413C>T (p.Pro138Leu)	ARSA	22	50627218	Metachromatic leukodystrophy, severe
NM_000487.5(ARSA):c.465+1G>A	ARSA	22	50627165	Metachromatic leukodystrophy;Metachromatic leukodystrophy, adult type;Metachromatic leukodystrophy, juvenile type;not provided
NM_000487.5(ARSA):c.1283C>T (p.Pro428Leu)	ARSA	22	50625392	Metachromatic leukodystrophy;Metachromatic leukodystrophy, adult type;Metachromatic leukodystrophy, juvenile type;not provided
NM_000487.5(ARSA):c.1210+1G>A	ARSA	22	50625578	Metachromatic leukodystrophy;Metachromatic leukodystrophy, juvenile type
NM_000487.5(ARSA):c.293C>T (p.Ser98Phe)	ARSA	22	50627338	Metachromatic leukodystrophy;Metachromatic leukodystrophy, late infantile
NM_000487.5(ARSA):c.763G>A (p.Glu255Lys)	ARSA	22	50626682	Metachromatic leukodystrophy;Metachromatic leukodystrophy, late infantile;not provided
NM_000487.5(ARSA):c.257G>A (p.Arg86Gln)	ARSA	22	50627374	Metachromatic leukodystrophy;Metachromatic leukodystrophy, late-onset
NM_000487.5(ARSA):c.1232C>T (p.Thr411Ile)	ARSA	22	50625443	Metachromatic leukodystrophy;Metachromatic leukodystrophy, mild
NM_000487.5(ARSA):c.739G>A (p.Gly247Arg)	ARSA	22	50626706	Metachromatic leukodystrophy;Metachromatic leukodystrophy, severe
NM_000487.5(ARSA):c.827C>T (p.Thr276Met)	ARSA	22	50626618	Metachromatic leukodystrophy;Metachromatic leukodystrophy, severe
NM_000487.5(ARSA):c.1114C>T (p.Arg372Trp)	ARSA	22	50625675	Metachromatic leukodystrophy;Metachromatic leukodystrophy, severe
NM_000487.5(ARSA):c.256C>T (p.Arg86Trp)	ARSA	22	50627375	Metachromatic leukodystrophy;not provided
NM_000487.5(ARSA):c.737G>A (p.Arg246His)	ARSA	22	50626708	Metachromatic leukodystrophy;not provided
NM_000487.5(ARSA):c.883G>A (p.Gly295Ser)	ARSA	22	50626250	Metachromatic leukodystrophy;not provided

NR_003051.3(RMRP):n.64C>T	RMRP	9	35657955	Metaphyseal chondrodysplasia, McKusick type
NM_000493.3(COL10A1):c.1953G>A (p.Trp651Ter)	COL10A1	6	116120163	Metaphyseal chondrodysplasia, Schmid type
NM_000493.3(COL10A1):c.52G>A (p.Gly18Arg)	COL10A1	6	116125441	Metaphyseal chondrodysplasia, Schmid type
NM_000493.3(COL10A1):c.53G>A (p.Gly18Glu)	COL10A1	6	116125440	Metaphyseal chondrodysplasia, Schmid type
NM_000493.3(COL10A1):c.1784G>A (p.Gly595Glu)	COL10A1	6	116120332	Metaphyseal chondrodysplasia, Schmid type
NM_000493.3(COL10A1):c.1832G>A (p.Trp611Ter)	COL10A1	6	116120284	Metaphyseal chondrodysplasia, Schmid type
NM_002427.3(MMP13):c.325C>T (p.Arg109Ter)	MMP13	11	102955289	Metaphyseal chondrodysplasia, Spahr type
NM_021625.4(TRPV4):c.2219C>T (p.Thr740Ile)	TRPV4	12	109786827	Metatrophic dysplasia;Neuromuscular Diseases;Skeletal dysplasia
NM_021625.4(TRPV4):c.2389G>A (p.Glu797Lys)	TRPV4	12	109784385	Metatrophic dysplasia;Neuromuscular Diseases;Skeletal dysplasia;Spondyloepiphyseal dysplasia Maroteaux type;Spondylometaphyseal dysplasia, Kozlowski type
NM_021625.4(TRPV4):c.2395C>T (p.Pro799Ser)	TRPV4	12	109784379	Metatrophic dysplasia;Skeletal dysplasia
NM_021625.4(TRPV4):c.266C>T (p.Thr89Ile)	TRPV4	12	109814531	Metatrophic dysplasia;Skeletal dysplasia
NM_021625.4(TRPV4):c.2396C>T (p.Pro799Leu)	TRPV4	12	109784378	Metatrophic dysplasia;Skeletal dysplasia;Spondyloepiphyseal dysplasia Maroteaux type
NM_000398.6(CYB5R3):c.655C>T (p.Arg219Ter)	CYB5R3	22	42623867	Methemoglobinemia type 2
NM_000398.6(CYB5R3):c.229C>T (p.Gln77Ter)	CYB5R3	22	42630986	Methemoglobinemia type 2
NM_000398.6(CYB5R3):c.478C>T (p.Arg160Ter)	CYB5R3	22	42627674	Methemoglobinemia type 2
NM_000398.6(CYB5R3):c.173G>A (p.Arg58Gln)	CYB5R3	22	42631431	Methemoglobinemia, type I
NM_000398.6(CYB5R3):c.316G>A (p.Val106Met)	CYB5R3	22	42630899	Methemoglobinemia, type I
NM_000398.6(CYB5R3):c.611G>A (p.Cys204Tyr)	CYB5R3	22	42627326	Methemoglobinemia, type I
NM_000398.6(CYB5R3):c.875G>A (p.Gly292Asp)	CYB5R3	22	42619804	Methemoglobinemia, type I
NM_000254.2(MTR):c.1753C>T (p.Arg585Ter)	MTR	1	236852578	METHYLCOBALAMIN DEFICIENCY, cblG TYPE
NM_005589.3(ALDH6A1):c.1336G>A (p.Gly446Arg)	ALDH6A1	14	74065249	Methylmalonate semialdehyde dehydrogenase deficiency
NM_005589.3(ALDH6A1):c.184C>T (p.Pro62Ser)	ALDH6A1	14	74072539	Methylmalonate semialdehyde dehydrogenase deficiency
NM_015506.2(MMACHC):c.609G>A (p.Trp203Ter)	MMACHC	1	45508975	Methylmalonic acidemia with homocystinuria
NM_015506.2(MMACHC):c.276G>A (p.Glu92=)	MMACHC	1	45507550	Methylmalonic acidemia with homocystinuria
NM_015506.2(MMACHC):c.464G>A (p.Gly155Glu)	MMACHC	1	45508830	Methylmalonic acidemia with homocystinuria
NM_015702.2(MMADHC):c.748C>T (p.Arg250Ter)	MMADHC	2	149570117	Methylmalonic acidemia with homocystinuria cblD
NM_015702.2(MMADHC):c.160C>T (p.Arg54Ter)	MMADHC	2	149579643	Methylmalonic acidemia with homocystinuria cblD;Methylmalonic aciduria, cblD type, variant 2
NM_015506.2(MMACHC):c.394C>T (p.Arg132Ter)	MMACHC	1	45508329	Methylmalonic acidemia with homocystinuria;not provided
NM_015506.2(MMACHC):c.331C>T (p.Arg111Ter)	MMACHC	1	45508266	Methylmalonic acidemia with homocystinuria;not provided
NM_015506.2(MMACHC):c.481C>T (p.Arg161Ter)	MMACHC	1	45508847	Methylmalonic acidemia with homocystinuria;not provided
NM_015506.2(MMACHC):c.608G>A (p.Trp203Ter)	MMACHC	1	45508974	Methylmalonic acidemia with homocystinuria;not provided
NM_052845.3(MMAB):c.556C>T (p.Arg186Trp)	MMAB	12	109561068	Methylmalonic acidemia;Methylmalonic aciduria cblB type;not provided
NM_172250.2(MMAA):c.283C>T (p.Gln95Ter)	MMAA	4	145639422	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.64C>T (p.Arg22Ter)	MMAA	4	145639203	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.161G>A (p.Trp54Ter)	MMAA	4	145639300	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.358C>T (p.Gln120Ter)	MMAA	4	145639497	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.397C>T (p.Gln133Ter)	MMAA	4	145639536	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.653G>A (p.Gly218Glu)	MMAA	4	145646076	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.733+1G>A	MMAA	4	145646157	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.1076G>A (p.Arg359Gln)	MMAA	4	145655253	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.562+1G>A	MMAA	4	145642486	Methylmalonic aciduria cblA type
NM_172250.2(MMAA):c.433C>T (p.Arg145Ter)	MMAA	4	145639572	Methylmalonic aciduria cblA type;not provided
NM_172250.2(MMAA):c.988C>T (p.Arg330Ter)	MMAA	4	145655165	Methylmalonic aciduria cblA type;not provided
NM_052845.3(MMAB):c.520_584del65	MMAB	12	109561040	Methylmalonic aciduria cblB type

NM_052845.3(MMAB):c.197_290del94	MMAB	12	109568770	Methylmalonic aciduria cblB type
NM_052845.3(MMAB):c.572G>A (p.Arg191Gln)	MMAB	12	109561052	Methylmalonic aciduria cblB type
NM_052845.3(MMAB):c.291-1G>A	MMAB	12	109565177	Methylmalonic aciduria cblB type
NM_052845.3(MMAB):c.568C>T (p.Arg190Cys)	MMAB	12	109561056	Methylmalonic aciduria cblB type;not provided
NM_052845.3(MMAB):c.700C>T (p.Gln234Ter)	MMAB	12	109557081	Methylmalonic aciduria cblB type;not provided
NM_052845.3(MMAB):c.569G>A (p.Arg190His)	MMAB	12	109561055	Methylmalonic aciduria cblB type;not provided
NM_052845.3(MMAB):c.571C>T (p.Arg191Trp)	MMAB	12	109561053	Methylmalonic aciduria cblB type;not provided
NM_000255.3(MUT):c.890C>T (p.Thr297Ile)	MUT	6	49456101	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.2179C>T (p.Arg727Ter)	MUT	6	49431802	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.2080C>T (p.Arg694Trp)	MUT	6	49435500	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.1280G>A (p.Gly427Asp)	MUT	6	49451518	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.607G>A (p.Gly203Arg)	MUT	6	49457837	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.19C>T (p.Gln7Ter)	MUT	6	49459448	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.1975C>T (p.Gln659Ter)	MUT	6	49435605	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.1655C>T (p.Ala552Val)	MUT	6	49444660	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.1277G>A (p.Gly426Glu)	MUT	6	49451521	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.1271C>T (p.Pro424Leu)	MUT	6	49451527	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.1084-1G>A	MUT	6	49451715	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.977G>A (p.Arg326Lys)	MUT	6	49453691	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.974G>A (p.Gly325Asp)	MUT	6	49453694	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.927G>A (p.Trp309Ter)	MUT	6	49453741	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.851G>A (p.Gly284Glu)	MUT	6	49456140	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.850G>A (p.Gly284Arg)	MUT	6	49456141	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.415G>A (p.Asp139Asn)	MUT	6	49458029	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.397G>A (p.Gly133Arg)	MUT	6	49458047	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.129G>A (p.Trp43Ter)	MUT	6	49459338	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.88C>T (p.Gln30Ter)	MUT	6	49459379	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.-39-1G>A	MUT	6	49459506	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.91C>T (p.Arg31Ter)	MUT	6	49459376	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency;not provided
NM_000255.3(MUT):c.1207C>T (p.Arg403Ter)	MUT	6	49451591	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency;not provided
NM_000255.3(MUT):c.280G>A (p.Gly94Arg)	MUT	6	49459187	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency;not provided
NM_000255.3(MUT):c.1663G>A (p.Ala555Thr)	MUT	6	49444652	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency;not provided
NM_000255.3(MUT):c.1106G>A (p.Arg369His)	MUT	6	49451692	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency;not provided
NM_000255.3(MUT):c.1105C>T (p.Arg369Cys)	MUT	6	49451693	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency;not provided
NM_000255.3(MUT):c.2200C>T (p.Gln734Ter)	MUT	6	49431781	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency;not provided
NM_000255.3(MUT):c.52C>T (p.Gln18Ter)	MUT	6	49459415	METHYLMALONIC ACIDURIA, mut(0) TYPE;Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.278G>A (p.Arg93His)	MUT	6	49459189	METHYLMALONIC ACIDURIA, mut(0) TYPE;Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.322C>T (p.Arg108Cys)	MUT	6	49459145	METHYLMALONIC ACIDURIA, mut(0) TYPE;Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.643G>A (p.Gly215Ser)	MUT	6	49457801	METHYLMALONIC ACIDURIA, mut(0) TYPE;Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
NM_000255.3(MUT):c.1867G>A (p.Gly623Arg)	MUT	6	49440295	METHYLMALONIC ACIDURIA, mut(0) TYPE;Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency;not provided

NM_032601.3(MCEE):c.139C>T (p.Arg47Ter)	MCEE	2	71124445	Methylmalonyl-CoA epimerase deficiency
NM_032601.3(MCEE):c.427C>T (p.Arg143Cys)	MCEE	2	71110074	Methylmalonyl-CoA epimerase deficiency
NM_139125.3(MASP1):c.1489C>T (p.His497Tyr)	MASP1	3	187236382	Michels syndrome
NM_139125.3(MASP1):c.1997G>A (p.Gly666Glu)	MASP1	3	187235874	Michels syndrome
NM_139125.3(MASP1):c.1779G>A (p.Trp593Ter)	MASP1	3	187236092	Michels syndrome
NM_006031.5(PCNT):c.5767C>T (p.Arg1923Ter)	PCNT	21	46411840	Microcephalic osteodysplastic primordial dwarfism type 2
NM_006031.5(PCNT):c.8752C>T (p.Arg2918Ter)	PCNT	21	46435904	Microcephalic osteodysplastic primordial dwarfism type 2
NM_006031.5(PCNT):c.1468C>T (p.Gln490Ter)	PCNT	21	46353115	Microcephalic osteodysplastic primordial dwarfism type 2
NM_006031.5(PCNT):c.8917C>T (p.Arg2973Ter)	PCNT	21	46436069	Microcephalic osteodysplastic primordial dwarfism type 2
NM_007174.2(CIT):c.1111+1G>A	CIT	12	119822819	MICROCEPHALY 17, PRIMARY, AUTOSOMAL RECESSIVE;Primary autosomal recessive microcephaly
NM_020461.3(TUBGCP6):c.2356C>T (p.Arg786Ter)	TUBGCP6	22	50222507	Microcephaly and chorioretinopathy with or without mental retardation
NM_020461.3(TUBGCP6):c.895C>T (p.Arg299Ter)	TUBGCP6	22	50240214	Microcephaly and chorioretinopathy with or without mental retardation
NM_020461.3(TUBGCP6):c.2215C>T (p.Arg739Ter)	TUBGCP6	22	50224196	Microcephaly with chorioretinopathy, autosomal recessive
NM_020461.3(TUBGCP6):c.3163C>T (p.His1055Tyr)	TUBGCP6	22	50221196	Microcephaly with chorioretinopathy, autosomal recessive
NM_004523.3(KIF11):c.1159C>T (p.Arg387Ter)	KIF11	10	92621415	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation
NM_004523.3(KIF11):c.2830C>T (p.Arg944Cys)	KIF11	10	92649894	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation
NM_004523.3(KIF11):c.790-1G>A	KIF11	10	92613376	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation
NM_004523.3(KIF11):c.2341C>T (p.Gln781Ter)	KIF11	10	92645436	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation
NM_002485.4(NBN):c.2165G>A (p.Trp722Ter)	NBN	8	89943272	Microcephaly, normal intelligence and immunodeficiency
NM_002485.4(NBN):c.1125G>A (p.Trp375Ter)	NBN	8	89955555	Microcephaly, normal intelligence and immunodeficiency
NM_005051.2(QARS):c.1207C>T (p.Arg403Trp)	QARS	3	49100049	Microcephaly, progressive, with seizures and cerebral and cerebellar atrophy
NM_005051.2(QARS):c.1543C>T (p.Arg515Trp)	QARS	3	49099415	Microcephaly, progressive, with seizures and cerebral and cerebellar atrophy;not provided
NM_001134665.2(TRMT10A):c.379C>T (p.Arg127Ter)	TRMT10A	4	99557386	Microcephaly, short stature, and impaired glucose metabolism 1
NM_152292.4(TRMT10A):c.616G>A (p.Gly206Arg)	TRMT10A	4	99553814	Microcephaly, short stature, and impaired glucose metabolism 1
NM_032833.4(PPP1R15B):c.1972C>T (p.Arg658Cys)	PPP1R15B	1	204406262	Microcephaly, short stature, and impaired glucose metabolism 2
NM_015114.2(ANKLE2):c.2344C>T (p.Gln782Ter)	ANKLE2	12	132729818	Microcephaly;Microcephaly 16, primary, autosomal recessive
NM_138615.2(DHX30):c.1478G>A (p.Arg493His)	DHX30	3	47846550	Microcephaly;Oculomotor apraxia;Seizures;Short stature;Strabismus;Unsteady gait
NM_201647.2(STAMBP):c.532C>T (p.Arg178Ter)	STAMBP	2	73847543	Microcephaly-capillary malformation syndrome
NM_201647.2(STAMBP):c.1270C>T (p.Arg424Ter)	STAMBP	2	73862254	Microcephaly-capillary malformation syndrome
NM_201647.2(STAMBP):c.112C>T (p.Arg38Cys)	STAMBP	2	73830968	Microcephaly-capillary malformation syndrome;not provided
NM_199355.3(ADAMTS18):c.97C>T (p.Gln33Ter)	ADAMTS18	16	77434499	Microcornea, myopic chorioretinal atrophy, and telecanthus
NM_004183.3(BEST1):c.715G>A (p.Val239Met)	BEST1	11	61958146	Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma
NM_153609.3(TMPRSS6):c.1582+1G>A	TMPRSS6	22	37073531	Microcytic anemia
NM_153609.3(TMPRSS6):c.1324G>A (p.Gly442Arg)	TMPRSS6	22	37075180	Microcytic anemia
NM_153609.3(TMPRSS6):c.1561G>A (p.Asp521Asn)	TMPRSS6	22	37073553	Microcytic anemia
NM_153609.3(TMPRSS6):c.1795C>T (p.Arg599Ter)	TMPRSS6	22	37070557	Microcytic anemia
NM_153609.3(TMPRSS6):c.1564G>A (p.Glu522Lys)	TMPRSS6	22	37073550	Microcytic anemia
NM_003106.3(SOX2):c.529C>T (p.Gln177Ter)	SOX2	3	181712889	Microphthalmia syndromic 3
NM_003106.3(SOX2):c.463C>T (p.Gln155Ter)	SOX2	3	181712823	Microphthalmia syndromic 3
NM_003106.3(SOX2):c.163C>T (p.Gln55Ter)	SOX2	3	181712523	Microphthalmia syndromic 3
NM_021728.3(OTX2):c.289C>T (p.Arg97Ter)	OTX2	14	56802340	Microphthalmia syndromic 5
NM_172337.2(OTX2):c.235G>A (p.Glu79Lys)	OTX2	14	56804202	Microphthalmia syndromic 5
NM_001202.5(BMP4):c.592C>T (p.Arg198Ter)	BMP4	14	53950667	Microphthalmia syndromic 6;Orofacial cleft 11
NM_001142617.1(STRA6):c.878C>T (p.Pro293Leu)	STRA6	15	74190889	Microphthalmia syndromic 9
NM_001142617.1(STRA6):c.1963C>T (p.Arg655Cys)	STRA6	15	74180121	Microphthalmia syndromic 9

NM_001142617.1(STRA6):c.1931C>T (p.Thr644Met)	STRA6	15	74180153	Microphthalmia syndromic 9	
NM_001142617.1(STRA6):c.269C>T (p.Pro90Leu)	STRA6	15	74196145	Microphthalmia syndromic 9	
NM_001142617.1(STRA6):c.69G>A (p.Trp23Ter)	STRA6	15	74202199	Microphthalmia syndromic 9	
NM_001142617.1(STRA6):c.1964G>A (p.Arg655His)	STRA6	15	74180120	Microphthalmia syndromic 9	
NM_001142617.1(STRA6):c.1521-1G>A	STRA6	15	74181459	Microphthalmia syndromic 9	
NM_182894.2(VSX2):c.599G>A (p.Arg200Gln)	VSX2	14	74259621	Microphthalmia, cataracts, and iris abnormalities	
NM_182894.2(VSX2):c.679C>T (p.Arg227Trp)	VSX2	14	74259701	Microphthalmia, isolated 2	
NM_013435.2(RAX):c.439C>T (p.Gln147Ter)	RAX	18	59272465	Microphthalmia, isolated 3	
NM_013435.2(RAX):c.575G>A (p.Arg192Gln)	RAX	18	59269470	Microphthalmia, isolated 3	
NM_031433.3(MFRP):c.201G>A (p.Trp67Ter)	MFRP	11	119346116	Microphthalmia, isolated 5	
NM_031433.3(MFRP):c.1549C>T (p.Arg517Trp)	MFRP	11	119341739	Microphthalmia, isolated 5	
NM_031433.3(MFRP):c.958C>T (p.Gln320Ter)	MFRP	11	119344332	Microphthalmia, isolated 5	
NM_031433.3(MFRP):c.523C>T (p.Gln175Ter)	MFRP	11	119345538	Microphthalmia, isolated 5;Nanophthalmos 2	
NM_001195129.1(PRSS56):c.958G>A (p.Gly320Arg)	PRSS56	2	232523524	Microphthalmia, isolated 6	
NM_001195129.1(PRSS56):c.709G>A (p.Gly237Arg)	PRSS56	2	232523062	Microphthalmia, isolated 6	
NM_001195129.1(PRSS56):c.1555G>A (p.Gly519Arg)	PRSS56	2	232525249	Microphthalmia, isolated 6	
NM_020634.2(GDF3):c.584G>A (p.Arg195Gln)	GDF3	12	7690389	Microphthalmia, isolated 7	
NM_000693.3(ALDH1A3):c.265C>T (p.Arg89Cys)	ALDH1A3	15	100887632	Microphthalmia, isolated 8	
NM_006744.3(RBP4):c.223G>A (p.Ala75Thr)	RBP4	10	93600692	Microphthalmia, isolated, with coloboma 10	
NM_006744.3(RBP4):c.217G>A (p.Ala73Thr)	RBP4	10	93600698	Microphthalmia, isolated, with coloboma 10	
NM_020634.2(GDF3):c.820C>T (p.Arg274Trp)	GDF3	12	7690153	Microphthalmia, isolated, with coloboma 6	
NM_005689.3(ABCB6):c.169G>A (p.Ala57Thr)	ABCB6	2	219218505	Microphthalmia, isolated, with coloboma 7	
NM_000965.4(RARB):c.355C>T (p.Arg119Ter)	RARB	3	25501230	Microphthalmia, syndromic 12	
NM_000965.4(RARB):c.1159C>T (p.Arg387Cys)	RARB	3	25596428	Microphthalmia, syndromic 12;not provided	
NM_006439.4(MAB21L2):c.152G>A (p.Arg51His)	MAB21L2	4	150583181	Microphthalmia, syndromic 14	
NM_006439.4(MAB21L2):c.151C>T (p.Arg51Cys)	MAB21L2	4	150583180	Microphthalmia, syndromic 14	
NM_006439.4(MAB21L2):c.145G>A (p.Glu49Lys)	MAB21L2	4	150583174	Microphthalmia, syndromic 14	
NM_006439.4(MAB21L2):c.740G>A (p.Arg247Gln)	MAB21L2	4	150583769	Microphthalmia, syndromic 14	
NM_000428.2(LTBP2):c.4855C>T (p.Gln1619Ter)	LTBP2	14	74503252	Microspherophakia	
NM_000428.2(LTBP2):c.4313G>A (p.Cys1438Tyr)	LTBP2	14	74505039	Microspherophakia	
NM_006735.3(HOXA2):c.703C>T (p.Gln235Ter)	HOXA2	7	27101154	Microtia, hearing impairment, and cleft palate	
NM_015365.2(AMMECR1):c.530G>A (p.Gly177Asp)	AMMECR1	X	110264543	MIDFACE HYPOPLASIA, HEARING IMPAIRMENT, ELLIPTOCYTOSIS, AND NEPHROCALCINOSIS	
NM_015365.2(AMMECR1):c.502C>T (p.Arg168Ter)	AMMECR1	X	110264571	MIDFACE HYPOPLASIA, HEARING IMPAIRMENT, ELLIPTOCYTOSIS, AND NEPHROCALCINOSIS	
NM_000702.3(ATP1A2):c.1643G>A (p.Arg548His)	ATP1A2	1	160130283	Migraine, familial basilar;not provided	
NM_001361.4(DHODH):c.1036C>T (p.Arg346Trp)	DHODH	16	72023536	Miller syndrome	
NM_001361.4(DHODH):c.454G>A (p.Gly152Arg)	DHODH	16	72017043	Miller syndrome	
NM_001361.4(DHODH):c.605G>A (p.Gly202Asp)	DHODH	16	72021211	Miller syndrome	
NM_001361.4(DHODH):c.730C>T (p.Arg244Trp)	DHODH	16	72022386	Miller syndrome	
NM_001361.4(DHODH):c.595C>T (p.Arg199Cys)	DHODH	16	72021201	Miller syndrome	
NM_001361.4(DHODH):c.403C>T (p.Arg135Cys)	DHODH	16	72014641	Miller syndrome;not provided	
NM_000540.2(RYR1):c.325C>T (p.Arg109Trp)	RYR1	19	38443612	Minicore myopathy with external ophthalmoplegia;not provided	
NM_000540.2(RYR1):c.10343C>T (p.Ser3448Phe)	RYR1	19	38523111	Minicore myopathy with external ophthalmoplegia;not provided	
NM_005215.3(DCC):c.1140+1G>A (p.V329GfsTer15)	DCC	18	53063460	Mirror movements 1	
NM_002875.4(RAD51):c.760C>T (p.Arg254Ter)	RAD51	15	40729620	Mirror movements 2	
NM_173560.3(RFX6):c.542G>A (p.Arg181Gln)	RFX6	6	116882404	Mitchell-Riley syndrome	

NM_173560.3(RFX6):c.2176C>T (p.Arg726Ter)	RFX6	6	116927317	Mitchell-Riley syndrome
NM_173560.3(RFX6):c.2596C>T (p.Arg866Ter)	RFX6	6	116928956	Mitchell-Riley syndrome
NM_005518.3(HMGCS2):c.634G>A (p.Gly212Arg)	HMGCS2	1	119759915	mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
NM_001166107.1(HMGCS2):c.1373G>A (p.Arg458His)	HMGCS2	1	119750830	mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
NM_001166107.1(HMGCS2):c.160G>A (p.Val54Met)	HMGCS2	1	119764571	mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
NM_017547.3(FOXRED1):c.694C>T (p.Gln232Ter)	FOXRED1	11	126275389	Mitochondrial complex I deficiency
NM_174889.4(NDUFAF2):c.139C>T (p.Arg47Ter)	NDUFAF2	5	61073136	Mitochondrial complex I deficiency
NM_004553.4(NDUFS6):c.344G>A (p.Cys115Tyr)	NDUFS6	5	1815885	Mitochondrial complex I deficiency
NM_004550.4(NDUFS2):c.683G>A (p.Arg228Gln)	NDUFS2	1	161209912	Mitochondrial complex I deficiency
NM_002495.3(NDUFS4):c.44G>A (p.Trp15Ter)	NDUFS4	5	53560706	Mitochondrial complex I deficiency
NM_002496.3(NDUFS8):c.254C>T (p.Pro85Leu)	NDUFS8	11	68033165	Mitochondrial complex I deficiency
NM_002496.3(NDUFS8):c.413G>A (p.Arg138His)	NDUFS8	11	68036293	Mitochondrial complex I deficiency
m.4810G>A	MT-ND2	MT	4810	Mitochondrial complex I deficiency
NM_007103.3(NDUFV1):c.175C>T (p.Arg59Ter)	NDUFV1	11	67608571	Mitochondrial complex I deficiency
NM_007103.3(NDUFV1):c.640G>A (p.Glu214Lys)	NDUFV1	11	67610510	Mitochondrial complex I deficiency
NM_005006.6(NDUFS1):c.721C>T (p.Arg241Trp)	NDUFS1	2	206146919	Mitochondrial complex I deficiency
NM_016013.3(NDUFAF1):c.631C>T (p.Arg211Cys)	NDUFAF1	15	41394987	Mitochondrial complex I deficiency
NM_017547.3(FOXRED1):c.1054C>T (p.Arg352Trp)	FOXRED1	11	126276476	Mitochondrial complex I deficiency
NM_002496.3(NDUFS8):c.229C>T (p.Arg77Trp)	NDUFS8	11	68033140	Mitochondrial complex I deficiency
NM_005006.6(NDUFS1):c.1669C>T (p.Arg557Ter)	NDUFS1	2	206130127	Mitochondrial complex I deficiency
NM_005006.6(NDUFS1):c.1222C>T (p.Arg408Cys)	NDUFS1	2	206141981	Mitochondrial complex I deficiency
NM_025152.2(NUBPL):c.693+1G>A	NUBPL	14	31826715	Mitochondrial complex I deficiency
NM_005006.6(NDUFS1):c.1855G>A (p.Asp619Asn)	NDUFS1	2	206127826	Mitochondrial complex I deficiency
NM_015965.6(NDUFA13):c.170G>A (p.Arg57His)	NDUFA13	19	19526257	Mitochondrial complex I deficiency
NM_018480.5(TMEM126B):c.397G>A (p.Asp133Asn)	TMEM126B	11	85634279	Mitochondrial complex I deficiency
NM_018480.5(TMEM126B):c.208C>T (p.Gln70Ter)	TMEM126B	11	85634090	Mitochondrial complex I deficiency
NM_002496.3(NDUFS8):c.236C>T (p.Pro79Leu)	NDUFS8	11	68033147	Mitochondrial complex I deficiency;not provided
NM_007103.3(NDUFV1):c.1268C>T (p.Thr423Met)	NDUFV1	11	67612225	Mitochondrial complex I deficiency;not provided
NM_019056.6(NDUFB11):c.391G>A (p.Glu131Lys)	NDUFB11	X	47142418	Mitochondrial complex I deficiency;not provided
NM_004168.3(SDHA):c.1571C>T (p.Ala524Val)	SDHA	5	251011	Mitochondrial complex II deficiency
NM_004168.3(SDHA):c.1526C>T (p.Ser509Leu)	SDHA	5	240451	Mitochondrial complex II deficiency
NM_003002.3(SDHD):c.205G>A (p.Glu69Lys)	SDHD	11	112088902	Mitochondrial complex II deficiency;not provided
NM_004168.3(SDHA):c.1054C>T (p.Arg352Ter)	SDHA	5	233635	Mitochondrial complex II deficiency;Paraganglioma 5
NM_004168.3(SDHA):c.1534C>T (p.Arg512Ter)	SDHA	5	240459	Mitochondrial complex II deficiency;Paraganglioma 5
NM_004168.3(SDHA):c.985C>T (p.Arg329Ter)	SDHA	5	233566	Mitochondrial complex II deficiency;Paraganglioma 5
NM_004168.3(SDHA):c.223C>T (p.Arg75Ter)	SDHA	5	224432	Mitochondrial complex II deficiency;Paraganglioma 5;not provided
NM_004328.4(BCS1L):c.830G>A (p.Ser277Asn)	BCS1L	2	218662620	Mitochondrial complex III deficiency
NM_004328.4(BCS1L):c.1057G>A (p.Val353Met)	BCS1L	2	218663183	Mitochondrial complex III deficiency
NM_004328.4(BCS1L):c.133C>T (p.Arg45Cys)	BCS1L	2	218661120	Mitochondrial complex III deficiency
NM_004328.4(BCS1L):c.547C>T (p.Arg183Cys)	BCS1L	2	218661845	Mitochondrial complex III deficiency
NM_004328.4(BCS1L):c.598C>T (p.Arg200Ter)	BCS1L	2	218661896	Mitochondrial complex III deficiency
NM_017775.3(TTC19):c.517C>T (p.Gln173Ter)	TTC19	17	16003885	Mitochondrial complex III deficiency, nuclear type 2
NM_017775.3(TTC19):c.829C>T (p.Gln277Ter)	TTC19	17	16025169	Mitochondrial complex III deficiency, nuclear type 2
NM_014402.4(UQCRQ):c.134C>T (p.Ser45Phe)	UQCRQ	5	132867015	Mitochondrial complex III deficiency, nuclear type 4;not provided
NM_003366.3(UQCRC2):c.547C>T (p.Arg183Trp)	UQCRC2	16	21965440	Mitochondrial complex III deficiency, nuclear type 5

NM_001916.4(CYC1):c.643C>T (p.Leu215Phe)	CYC1	8	144096615	Mitochondrial complex III deficiency, nuclear type 6;not provided
NM_181705.3(LYRM7):c.73G>A (p.Asp25Asn)	LYRM7	5	131180149	Mitochondrial complex III deficiency, nuclear type 8
NM_181705.3(LYRM7):c.214C>T (p.Gln72Ter)	LYRM7	5	131187079	Mitochondrial complex III deficiency, nuclear type 8
NM_001001937.1(ATP5A1):c.985C>T (p.Arg329Cys)	ATP5A1	18	46087199	Mitochondrial complex v (atp synthase) deficiency, nuclear type 4
NM_017909.3(RMND1):c.1303C>T (p.Leu435Phe)	RMND1	6	151405734	Mitochondrial diseases
NM_017909.3(RMND1):c.830+1G>A	RMND1	6	151427481	Mitochondrial diseases
NM_017909.3(RMND1):c.631G>A (p.Val211Met)	RMND1	6	151433213	Mitochondrial diseases
NM_017909.3(RMND1):c.565C>T (p.Gln189Ter)	RMND1	6	151436494	Mitochondrial diseases
NM_002693.2(POLG):c.695G>A (p.Arg232His)	POLG	15	89330241	Mitochondrial diseases;not provided
m.8313G>A	MT-TK	MT	8313	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001113755.2(TYMP):c.433G>A (p.Gly145Arg)	TYMP	22	50528595	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001113755.2(TYMP):c.457G>A (p.Gly153Ser)	TYMP	22	50528571	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001113755.2(TYMP):c.131G>A (p.Arg44Gln)	TYMP	22	50529579	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257989.1(TYMP):c.228G>A (p.Met76Ile)	TYMP	22	50529325	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257989.1(TYMP):c.328C>T (p.Gln110Ter)	TYMP	22	50529225	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257989.1(TYMP):c.340G>A (p.Asp114Asn)	TYMP	22	50529213	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257989.1(TYMP):c.605G>A (p.Arg202Lys)	TYMP	22	50527629	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257988.1(TYMP):c.1159G>A (p.Gly387Ser)	TYMP	22	50526246	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257989.1(TYMP):c.1175G>A (p.Gly392Asp)	TYMP	22	50526141	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257989.1(TYMP):c.1297G>A (p.Gly433Ser)	TYMP	22	50526019	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257989.1(TYMP):c.1326G>A (p.Trp442Ter)	TYMP	22	50525908	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001257989.1(TYMP):c.1174G>A (p.Gly392Ser)	TYMP	22	50526142	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001953.4(TYMP):c.1300+1G>A	TYMP	22	50526000	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001953.4(TYMP):c.1301-1G>A	TYMP	22	50525919	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
NM_001953.4(TYMP):c.622G>A (p.Val208Met)	TYMP	22	50527612	Mitochondrial DNA depletion syndrome 1 (MNGIE type);not provided
NM_001310338.1(MGME1):c.456G>A (p.Trp152Ter)	MGME1	20	17970315	Mitochondrial DNA depletion syndrome 11
NM_001151.3(SLC25A4):c.111+1G>A	SLC25A4	4	185143484	Mitochondrial DNA depletion syndrome 12 (cardiomyopathic type)
NM_001151.3(SLC25A4):c.239G>A (p.Arg80His)	SLC25A4	4	185144891	MITOCHONDRIAL DNA DEPLETION SYNDROME 12A (CARDIOMYOPATHIC TYPE), AUTOSOMAL DOMINANT;not provided
NM_012160.4(FBXL4):c.1555C>T (p.Gln519Ter)	FBXL4	6	98875562	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)
NM_012160.4(FBXL4):c.1303C>T (p.Arg435Ter)	FBXL4	6	98899282	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)
NM_012160.4(FBXL4):c.64C>T (p.Arg22Ter)	FBXL4	6	98926925	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)
NM_003201.2(TFAM):c.533C>T (p.Pro178Leu)	TFAM	10	58390856	MITOCHONDRIAL DNA DEPLETION SYNDROME 15 (HEPATOCEREBRAL TYPE);Mitochondrial DNA depletion syndrome, hepatocerebral form
NM_004614.4(TK2):c.133C>T (p.Gln45Ter)	TK2	16	66549001	Mitochondrial DNA depletion syndrome 2
NM_004614.4(TK2):c.191C>T (p.Thr64Met)	TK2	16	66541919	Mitochondrial DNA depletion syndrome 2
NM_004614.4(TK2):c.268C>T (p.Arg90Cys)	TK2	16	66536981	Mitochondrial DNA depletion syndrome 2
NM_004614.4(TK2):c.373C>T (p.Gln125Ter)	TK2	16	66531382	Mitochondrial DNA depletion syndrome 2
NM_004614.4(TK2):c.388C>T (p.Arg130Trp)	TK2	16	66529055	Mitochondrial DNA depletion syndrome 2
NM_004614.4(TK2):c.389G>A (p.Arg130Gln)	TK2	16	66529054	Mitochondrial DNA depletion syndrome 2
NM_004614.4(TK2):c.575G>A (p.Arg192Lys)	TK2	16	66517179	Mitochondrial DNA depletion syndrome 2
NM_004614.4(TK2):c.760C>T (p.Arg254Ter)	TK2	16	66512006	Mitochondrial DNA depletion syndrome 2
NM_004614.4(TK2):c.323C>T (p.Thr108Met)	TK2	16	66531432	Mitochondrial DNA depletion syndrome 2;not provided
NM_004614.4(TK2):c.416C>T (p.Ala139Val)	TK2	16	66529027	Mitochondrial DNA depletion syndrome 2;not provided

NM_004614.4(TK2):c.547C>T (p.Arg183Trp)	TK2	16	66517207	Mitochondrial DNA depletion syndrome 2;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3
NM_002693.2(POLG):c.679C>T (p.Arg227Trp)	POLG	15	89330257	Mitochondrial DNA depletion syndrome 4B, MNGIE type;not provided
NM_002693.2(POLG):c.3218C>T (p.Pro1073Leu)	POLG	15	89318986	Mitochondrial DNA depletion syndrome 4B, MNGIE type;Progressive sclerosing poliodystrophy;not provided
NM_003850.2(SUCLA2):c.534+1G>A	SUCLA2	13	47988540	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
NM_003850.2(SUCLA2):c.352G>A (p.Gly118Arg)	SUCLA2	13	47988901	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
NM_003850.2(SUCLA2):c.850C>T (p.Arg284Cys)	SUCLA2	13	47954510	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
NM_003850.2(SUCLA2):c.751G>A (p.Asp251Asn)	SUCLA2	13	47968646	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
NM_021830.4(TWNK):c.952G>A (p.Ala318Thr)	TWNK	10	100989162	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)
NM_021830.4(TWNK):c.1287C>T (p.Ala429=)	TWNK	10	100989687	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)
NM_021830.4(TWNK):c.1370C>T (p.Thr457Ile)	TWNK	10	100989770	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type);Mitochondrial DNA-depletion syndrome 3, hepatocerebral
NM_015713.4(RRM2B):c.329G>A (p.Arg110His)	RRM2B	8	102225011	Mitochondrial DNA depletion syndrome 8B (MNGIE type);RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.362G>A (p.Arg121His)	RRM2B	8	102224978	Mitochondrial DNA depletion syndrome 8B (MNGIE type);RRM2B-related mitochondrial disease
NM_003849.3(SUCLG1):c.448C>T (p.Gln150Ter)	SUCLG1	2	84441330	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic with methylmalonic aciduria)
NM_015713.4(RRM2B):c.850C>T (p.Gln284Ter)	RRM2B	8	102212829	Mitochondrial DNA depletion syndrome, encephalomyopathic form, with renal tubulopathy;RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.580G>A (p.Glu194Lys)	RRM2B	8	102218918	Mitochondrial DNA depletion syndrome, encephalomyopathic form, with renal tubulopathy;RRM2B-related mitochondrial disease
NM_080916.2(DGUOK):c.313C>T (p.Arg105Ter)	DGUOK	2	73946776	Mitochondrial DNA-depletion syndrome 3, hepatocerebral
NM_080916.2(DGUOK):c.425G>A (p.Arg142Lys)	DGUOK	2	73946888	Mitochondrial DNA-depletion syndrome 3, hepatocerebral
NM_080916.2(DGUOK):c.679G>A (p.Glu227Lys)	DGUOK	2	73957212	Mitochondrial DNA-depletion syndrome 3, hepatocerebral
m.12315G>A	MT-TL2	MT	12315	Mitochondrial encephalomyopathy
m.15242G>A	MT-CYB	MT	15242	Mitochondrial encephalomyopathy
m.5556G>A	MT-TW	MT	5556	Mitochondrial encephalomyopathy
m.5521G>A	MT-TW	MT	5521	Mitochondrial myopathy
m.5591G>A	MT-TA	MT	5591	Mitochondrial myopathy
NC_012920.1:m.5610G>A	MT-TA	MT	5610	Mitochondrial myopathy
NC_012920.1:m.5631G>A	MT-TA	MT	5631	Mitochondrial myopathy
NM_005888.3(SLC25A3):c.215G>A (p.Gly72Glu)	SLC25A3	12	98595490	Mitochondrial phosphate carrier deficiency
NM_016098.3(MPC1):c.289C>T (p.Arg97Trp)	MPC1	6	166365990	Mitochondrial pyruvate carrier deficiency
NM_004092.3(ECHS1):c.413C>T (p.Ala138Val)	ECHS1	10	133369905	Mitochondrial short-chain enoyl-coa hydratase 1 deficiency
NM_000182.4(HADHA):c.1132C>T (p.Gln378Ter)	HADHA	2	26204150	Mitochondrial trifunctional protein deficiency
NM_000182.4(HADHA):c.180+1G>A	HADHA	2	26238933	Mitochondrial trifunctional protein deficiency
NM_000182.4(HADHA):c.1678C>T (p.Arg560Ter)	HADHA	2	26194581	Mitochondrial trifunctional protein deficiency
NM_000182.4(HADHA):c.871C>T (p.Arg291Ter)	HADHA	2	26214490	Mitochondrial trifunctional protein deficiency
NM_000183.2(HADHB):c.182G>A (p.Arg61His)	HADHB	2	26263452	Mitochondrial trifunctional protein deficiency
NM_000183.2(HADHB):c.1331G>A (p.Arg444Lys)	HADHB	2	26285513	Mitochondrial trifunctional protein deficiency
NM_000183.2(HADHB):c.1175C>T (p.Ala392Val)	HADHB	2	26284908	Mitochondrial trifunctional protein deficiency
NM_000183.2(HADHB):c.740G>A (p.Arg247His)	HADHB	2	26279244	Mitochondrial trifunctional protein deficiency;not provided

NM_003737.3(DCHS1):c.7538G>A (p.Arg2513His)	DCHS1	11	6624138	Mitral valve prolapse 2
NM_003737.3(DCHS1):c.6988C>T (p.Arg2330Cys)	DCHS1	11	6625356	Mitral valve prolapse 2
NM_003494.3(DYSF):c.1129C>T (p.Arg377Ter)	DYSF	2	71526295	Miyoshi muscular dystrophy 1;not provided
NM_213599.2(ANO5):c.2235+1G>A	ANO5	11	22272990	Miyoshi muscular dystrophy 3
NM_004085.3(TIMM8A):c.238C>T (p.Arg80Ter)	TIMM8A	X	101346555	Mohr-Tranebjaerg syndrome
NM_004085.3(TIMM8A):c.112C>T (p.Gln38Ter)	TIMM8A	X	101348553	Mohr-Tranebjaerg syndrome
NM_001075098.3(MOCS1):c.956G>A (p.Arg319Gln)	MOCS1	6	39912289	Molybdenum cofactor deficiency, complementation group A
NM_001075098.3(MOCS1):c.418+1G>A	MOCS1	6	39925677	Molybdenum cofactor deficiency, complementation group A
NM_001075098.3(MOCS1):c.217C>T (p.Arg73Trp)	MOCS1	6	39927362	Molybdenum cofactor deficiency, complementation group A
NM_004531.4(MOCS2):c.502G>A (p.Glu168Lys)	MOCS2	5	53098667	Molybdenum cofactor deficiency, complementation group B
NM_004531.4(MOCS2):c.3G>A (p.Met1Ile)	MOCS2	5	53107172	Molybdenum cofactor deficiency, complementation group B
NM_176806.3(MOCS2):c.16C>T (p.Gln6Ter)	MOCS2	5	53109714	Molybdenum cofactor deficiency, complementation group B
NM_000240.3(MAOA):c.886C>T (p.Gln296Ter)	MAOA	X	43731784	Monoamine oxidase A deficiency
NM_000240.3(MAOA):c.133C>T (p.Arg45Trp)	MAOA	X	43683572	Monoamine oxidase A deficiency
NM_003051.3(SLC16A1):c.586C>T (p.Arg196Ter)	SLC16A1	1	112917820	Monocarboxylate transporter 1 deficiency, autosomal dominant
NM_003051.3(SLC16A1):c.938G>A (p.Arg313Gln)	SLC16A1	1	112917468	Monocarboxylate transporter 1 deficiency, autosomal dominant
NM_003051.3(SLC16A1):c.937C>T (p.Arg313Ter)	SLC16A1	1	112917469	Monocarboxylate transporter 1 deficiency, autosomal recessive
NM_003051.3(SLC16A1):c.982C>T (p.Arg328Ter)	SLC16A1	1	112917424	Monocarboxylate transporter 1 deficiency, autosomal recessive
NM_000162.3(GCK):c.680-1G>A	GCK	7	44147834	Monogenic diabetes
NM_032898.4(CEP19):c.244C>T (p.Arg82Ter)	CEP19	3	196707811	Morbid obesity and spermatogenic failure
NM_019892.5(INPP5E):c.1879C>T (p.Gln627Ter)	INPP5E	9	136429731	MORM syndrome
NM_001211.5(BUB1B):c.2308C>T (p.Arg770Ter)	BUB1B	15	40210133	Mosaic variegated aneuploidy syndrome
NM_014679.4(CEP57):c.241C>T (p.Arg81Ter)	CEP57	11	95812970	Mosaic variegated aneuploidy syndrome 2
NM_001211.5(BUB1B):c.580C>T (p.Arg194Ter)	BUB1B	15	40176672	Mosaic variegated aneuploidy syndrome;Premature chromatid separation trait
NM_001211.5(BUB1B):c.2530C>T (p.Leu844Phe)	BUB1B	15	40212643	Mosaic variegated aneuploidy syndrome;Premature chromatid separation trait
NM_001211.5(BUB1B):c.2441G>A (p.Arg814His)	BUB1B	15	40212554	Mosaic variegated aneuploidy syndrome;Premature chromatid separation trait
NM_014795.3(ZEB2):c.-69-1G>A	ZEB2	2	144517420	Mowat-Wilson syndrome
NM_014795.3(ZEB2):c.1381C>T (p.Gln461Ter)	ZEB2	2	144399806	Mowat-Wilson syndrome
NM_014795.3(ZEB2):c.805C>T (p.Gln269Ter)	ZEB2	2	144403918	Mowat-Wilson syndrome
NM_014795.3(ZEB2):c.1956C>T (p.Tyr652=)	ZEB2	2	144399231	Mowat-Wilson syndrome
NM_014795.3(ZEB2):c.823C>T (p.Gln275Ter)	ZEB2	2	144401292	Mowat-Wilson syndrome
NM_014795.3(ZEB2):c.1027C>T (p.Arg343Ter)	ZEB2	2	144400160	Mowat-Wilson syndrome
NM_014795.3(ZEB2):c.2701C>T (p.Gln901Ter)	ZEB2	2	144398486	Mowat-Wilson syndrome
NM_014795.3(ZEB2):c.2083C>T (p.Arg695Ter)	ZEB2	2	144399104	Mowat-Wilson syndrome;not provided
NM_014795.3(ZEB2):c.2761C>T (p.Arg921Ter)	ZEB2	2	144398426	Mowat-Wilson syndrome;not provided
NM_014795.3(ZEB2):c.904C>T (p.Arg302Ter)	ZEB2	2	144401211	Mowat-Wilson syndrome;not provided
NM_014795.3(ZEB2):c.1102C>T (p.Gln368Ter)	ZEB2	2	144400085	Mowat-Wilson syndrome;not provided
NM_000856.5(GUCY1A3):c.1086+1G>A	GUCY1A3	4	155711252	Moyamoya disease 6 with achalasia
NM_000856.5(GUCY1A3):c.1045C>T (p.Arg349Ter)	GUCY1A3	4	155711210	Moyamoya disease 6 with achalasia
NM_002437.4(MPV17):c.293C>T (p.Pro98Leu)	MPV17	2	27312576	MPV17-Related Disorders;Navajo neurohepatopathy;not provided
NM_024312.4(GNPTAB):c.771G>A (p.Leu257=)	GNPTAB	12	101780152	Mucolipidosis III alpha/beta, atypical;Pseudo-Hurler polydystrophy
NM_032520.4(GNPTG):c.333G>A (p.Trp111Ter)	GNPTG	16	1362053	Mucolipidosis III Gamma
NM_032520.4(GNPTG):c.316G>A (p.Gly106Ser)	GNPTG	16	1361954	Mucolipidosis III Gamma
NM_032520.4(GNPTG):c.196C>T (p.Arg66Ter)	GNPTG	16	1361760	Mucolipidosis III Gamma
NM_032520.4(GNPTG):c.857C>T (p.Thr286Met)	GNPTG	16	1363030	Mucolipidosis III Gamma

NM_000203.4(IDUA):c.590-7G>A	IDUA	4	1001672	Mucopolysaccharidosis type 1
NM_000203.4(IDUA):c.223G>A (p.Ala75Thr)	IDUA	4	987873	Mucopolysaccharidosis type 1
NM_000046.3(ARSB):c.1214G>A (p.Cys405Tyr)	ARSB	5	78781974	Mucopolysaccharidosis type VI;Mucopolysaccharidosis, type vi, severe
NM_000046.3(ARSB):c.284G>A (p.Arg95Gln)	ARSB	5	78984965	Mucopolysaccharidosis type VI;not provided
NM_000181.3(GUSB):c.1856C>T (p.Ala619Val)	GUSB	7	65960997	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.646C>T (p.Arg216Trp)	GUSB	7	65979477	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.1061C>T (p.Ala354Val)	GUSB	7	65974923	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.1831C>T (p.Arg611Trp)	GUSB	7	65961022	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.442C>T (p.Pro148Ser)	GUSB	7	65979866	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.1521G>A (p.Trp507Ter)	GUSB	7	65967863	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.1616_1653del38 (p.Ser539Argfs*8)	GUSB	7	65967767	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.1338G>A (p.Trp446Ter)	GUSB	7	65974348	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.530C>T (p.Thr177Ile)	GUSB	7	65979778	Mucopolysaccharidosis type VII
NM_000181.3(GUSB):c.1144C>T (p.Arg382Cys)	GUSB	7	65974626	Mucopolysaccharidosis type VII;Non-immune hydrops fetalis
NM_000181.3(GUSB):c.1069C>T (p.Arg357Ter)	GUSB	7	65974701	Mucopolysaccharidosis type VII;Non-immune hydrops fetalis
NM_000181.3(GUSB):c.526C>T (p.Leu176Phe)	GUSB	7	65979782	Mucopolysaccharidosis type VII;not provided
NM_000181.3(GUSB):c.1084G>A (p.Asp362Asn)	GUSB	7	65974686	Mucopolysaccharidosis type VII;not provided
NM_000181.3(GUSB):c.866G>A (p.Trp289Ter)	GUSB	7	65976061	Mucopolysaccharidosis type VII;not provided
NM_000203.4(IDUA):c.1091C>T (p.Thr364Met)	IDUA	4	1002387	Mucopolysaccharidosis, MPS-I-H/S
NM_000202.7(IDS):c.1327C>T (p.Arg443Ter)	IDS	X	149483072	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.514C>T (p.Arg172Ter)	IDS	X	149498301	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.1122C>T (p.Gly374=)	IDS	X	149486983	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.1425G>A (p.Trp475Ter)	IDS	X	149482974	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.1393C>T (p.Gln465Ter)	IDS	X	149483006	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.1181-1G>A	IDS	X	149483219	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.935G>A (p.Gly312Asp)	IDS	X	149490385	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.708+1G>A	IDS	X	149498106	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.592G>A (p.Asp198Asn)	IDS	X	149498223	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.508-1G>A	IDS	X	149498308	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.469C>T (p.Pro157Ser)	IDS	X	149500987	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.401G>A (p.Gly134Glu)	IDS	X	149503329	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.1591C>T (p.Gln531Ter)	IDS	X	149482808	Mucopolysaccharidosis, MPS-II
NM_000202.7(IDS):c.1402C>T (p.Arg468Trp)	IDS	X	149482997	Mucopolysaccharidosis, MPS-II;Mucopolysaccharidosis, type II, mild form
NM_000202.7(IDS):c.1403G>A (p.Arg468Gln)	IDS	X	149482996	Mucopolysaccharidosis, MPS-II;Mucopolysaccharidosis, type II, severe form;not provided
NM_000202.7(IDS):c.998C>T (p.Ser333Leu)	IDS	X	149490322	Mucopolysaccharidosis, MPS-II;not provided
NM_000202.7(IDS):c.253G>A (p.Ala85Thr)	IDS	X	149503477	Mucopolysaccharidosis, MPS-II;not provided
NM_000202.7(IDS):c.262C>T (p.Arg88Cys)	IDS	X	149503468	Mucopolysaccharidosis, MPS-II;not provided
NM_000199.3(SGSH):c.383C>T (p.Pro128Leu)	SGSH	17	80214738	Mucopolysaccharidosis, MPS-III-A
NM_000199.3(SGSH):c.449G>A (p.Arg150Gln)	SGSH	17	80214672	Mucopolysaccharidosis, MPS-III-A
NM_024312.4(GNPTAB):c.3435-1G>A	GNPTAB	12	101753540	Mucopolysaccharidosis, MPS-III-A
NM_024312.4(GNPTAB):c.832C>T (p.Gln278Ter)	GNPTAB	12	101771097	Mucopolysaccharidosis, MPS-III-A
NM_000199.3(SGSH):c.220C>T (p.Arg74Cys)	SGSH	17	80217061	Mucopolysaccharidosis, MPS-III-A;not provided
NM_000199.3(SGSH):c.1298G>A (p.Arg433Gln)	SGSH	17	80210663	Mucopolysaccharidosis, MPS-III-A;not provided
NM_000199.3(SGSH):c.1105G>A (p.Glu369Lys)	SGSH	17	80210856	Mucopolysaccharidosis, MPS-III-A;not provided

NM_000199.3(SGSH):c.877C>T (p.Pro293Ser)	SGSH	17	80212143	Mucopolysaccharidosis, MPS-III-A;not provided
NM_000199.3(SGSH):c.734G>A (p.Arg245His)	SGSH	17	80213815	Mucopolysaccharidosis, MPS-III-A;Sanfilippo syndrome;not provided
NM_000263.3(NAGLU):c.1928G>A (p.Arg643His)	NAGLU	17	42543934	Mucopolysaccharidosis, MPS-III-B
NM_000263.3(NAGLU):c.1927C>T (p.Arg643Cys)	NAGLU	17	42543933	Mucopolysaccharidosis, MPS-III-B
NM_000263.3(NAGLU):c.1562C>T (p.Pro521Leu)	NAGLU	17	42543568	Mucopolysaccharidosis, MPS-III-B
NM_000263.3(NAGLU):c.1444C>T (p.Arg482Trp)	NAGLU	17	42543450	Mucopolysaccharidosis, MPS-III-B
NM_000263.3(NAGLU):c.700C>T (p.Arg234Cys)	NAGLU	17	42538691	Mucopolysaccharidosis, MPS-III-B
NM_000263.3(NAGLU):c.1949G>A (p.Gly650Glu)	NAGLU	17	42543955	Mucopolysaccharidosis, MPS-III-B
NM_000263.3(NAGLU):c.694C>T (p.Gln232Ter)	NAGLU	17	42538685	Mucopolysaccharidosis, MPS-III-B
NM_000263.3(NAGLU):c.2021G>A (p.Arg674His)	NAGLU	17	42544027	Mucopolysaccharidosis, MPS-III-B;not provided
NM_000263.3(NAGLU):c.1876C>T (p.Arg626Ter)	NAGLU	17	42543882	Mucopolysaccharidosis, MPS-III-B;not provided
NM_000263.3(NAGLU):c.1694G>A (p.Arg565Gln)	NAGLU	17	42543700	Mucopolysaccharidosis, MPS-III-B;not provided
NM_000263.3(NAGLU):c.1693C>T (p.Arg565Trp)	NAGLU	17	42543699	Mucopolysaccharidosis, MPS-III-B;Sanfilippo syndrome
NM_000263.3(NAGLU):c.889C>T (p.Arg297Ter)	NAGLU	17	42541074	Mucopolysaccharidosis, MPS-III-B;Sanfilippo syndrome;not provided
NM_152419.2(HGSNAT):c.493+1G>A	HGSNAT	8	43159045	Mucopolysaccharidosis, MPS-III-C
NM_152419.2(HGSNAT):c.848C>T (p.Pro283Leu)	HGSNAT	8	43173740	Mucopolysaccharidosis, MPS-III-C
NM_152419.2(HGSNAT):c.1030C>T (p.Arg344Cys)	HGSNAT	8	43182162	Mucopolysaccharidosis, MPS-III-C
NM_152419.2(HGSNAT):c.1553C>T (p.Ser518Phe)	HGSNAT	8	43197682	Mucopolysaccharidosis, MPS-III-C
NM_152419.2(HGSNAT):c.518G>A (p.Gly173Asp)	HGSNAT	8	43161462	Mucopolysaccharidosis, MPS-III-C
NM_152419.2(HGSNAT):c.1360C>T (p.Gln454Ter)	HGSNAT	8	43192413	Mucopolysaccharidosis, MPS-III-C
NM_152419.2(HGSNAT):c.2344+1G>A	HGSNAT	8	43147064	Mucopolysaccharidosis, MPS-III-C;not provided
NM_002076.3(GNS):c.1063C>T (p.Arg355Ter)	GNS	12	64737039	Mucopolysaccharidosis, MPS-III-D
NM_002076.3(GNS):c.1168C>T (p.Gln390Ter)	GNS	12	64728988	Mucopolysaccharidosis, MPS-III-D
NM_000512.4(GALNS):c.1417C>T (p.Gln473Ter)	GALNS	16	88818072	Mucopolysaccharidosis, MPS-IV-A
NM_000512.4(GALNS):c.776G>A (p.Arg259Gln)	GALNS	16	88835335	Mucopolysaccharidosis, MPS-IV-A
NM_000512.4(GALNS):c.871G>A (p.Ala291Thr)	GALNS	16	88835240	Mucopolysaccharidosis, MPS-IV-A
NM_000512.4(GALNS):c.485C>T (p.Ser162Phe)	GALNS	16	88837703	Mucopolysaccharidosis, MPS-IV-A
NM_000512.4(GALNS):c.689G>A (p.Trp230Ter)	GALNS	16	88835794	Mucopolysaccharidosis, MPS-IV-A
NM_000512.4(GALNS):c.178G>A (p.Asp60Asn)	GALNS	16	88842772	Mucopolysaccharidosis, MPS-IV-A
NM_000512.4(GALNS):c.1019G>A (p.Gly340Asp)	GALNS	16	88826822	Mucopolysaccharidosis, MPS-IV-A
NM_000512.4(GALNS):c.139G>A (p.Gly47Arg)	GALNS	16	88842811	Mucopolysaccharidosis, MPS-IV-A
NM_000512.4(GALNS):c.1156C>T (p.Arg386Cys)	GALNS	16	88824853	Mucopolysaccharidosis, MPS-IV-A;not provided
NM_000512.4(GALNS):c.1175C>T (p.Ala392Val)	GALNS	16	88824834	Mucopolysaccharidosis, MPS-IV-A;not provided
NM_000512.4(GALNS):c.1559G>A (p.Trp520Ter)	GALNS	16	88814449	Mucopolysaccharidosis, MPS-IV-A;not provided
NM_000512.4(GALNS):c.463G>A (p.Gly155Arg)	GALNS	16	88837725	Mucopolysaccharidosis, MPS-IV-A;not provided
NM_000512.4(GALNS):c.477G>A (p.Trp159Ter)	GALNS	16	88837711	Mucopolysaccharidosis, MPS-IV-A;not provided
NM_000404.3(GLB1):c.1444C>T (p.Arg482Cys)	GLB1	3	33016744	Mucopolysaccharidosis, MPS-IV-B
NM_000404.3(GLB1):c.1313G>A (p.Gly438Glu)	GLB1	3	33018482	Mucopolysaccharidosis, MPS-IV-B
NM_022916.4(VPS33A):c.1492C>T (p.Arg498Trp)	VPS33A	12	122232917	MUCOPOLYSACCHARIDOSIS-PLUS SYNDROME
NM_015294.4(TRIM37):c.860G>A (p.Ser287Asn)	TRIM37	17	59064355	Mulibrey nanism syndrome
NM_030761.4(WNT4):c.247C>T (p.Arg83Trp)	WNT4	1	22129682	Mullerian aplasia and hyperandrogenism
NM_002438.3(MRC1):c.1186G>A (p.Gly396Ser)	MRC1	10	17849701	Multibacillary leprosy
NM_005461.4(MAFB):c.209C>T (p.Ser70Leu)	MAFB	20	40688642	Multicentric osteolysis nephropathy
NM_005461.4(MAFB):c.211C>T (p.Pro71Ser)	MAFB	20	40688640	Multicentric osteolysis nephropathy
NM_005461.4(MAFB):c.212C>T (p.Pro71Leu)	MAFB	20	40688639	Multicentric osteolysis nephropathy

NM_005461.4(MAFB):c.161C>T (p.Ser54Leu)	MAFB	20	40688690	Multicentric osteolysis nephropathy
NM_004530.5(MMP2):c.302G>A (p.Arg101His)	MMP2	16	55483057	Multicentric osteolysis, nodulosis and arthropathy
NM_004530.5(MMP2):c.1210G>A (p.Glu404Lys)	MMP2	16	55491830	Multicentric osteolysis, nodulosis and arthropathy
NM_001080508.2(TBX18):c.1570C>T (p.His524Tyr)	TBX18	6	84736939	Multicystic renal dysplasia, bilateral
NM_000267.3(NF1):c.4537C>T (p.Arg1513Ter)	NF1	17	31261733	Multiple cafe-au-lait spots;Neurofibromatosis, type 1;Subcutaneous neurofibromas;Thoracic scoliosis;not provided
NM_001042492.2(NF1):c.6904C>T (p.Gln2302Ter)	NF1	17	31338788	Multiple cafe-au-lait spots;Subcutaneous neurofibromas
NM_176787.4(PIGN):c.2126G>A (p.Arg709Gln)	PIGN	18	62095902	Multiple congenital anomalies-hypotonia-seizures syndrome 1
NM_176787.4(PIGN):c.963G>A (p.Gln321=)	PIGN	18	62143306	Multiple congenital anomalies-hypotonia-seizures syndrome 1
NM_176787.4(PIGN):c.1434+5G>A	PIGN	18	62113129	Multiple congenital anomalies-hypotonia-seizures syndrome 1
NM_176787.4(PIGN):c.1966C>T (p.Gln656Ter)	PIGN	18	62102796	Multiple congenital anomalies-hypotonia-seizures syndrome 1
NM_002641.3(PIGA):c.1234C>T (p.Arg412Ter)	PIGA	X	15321727	Multiple congenital anomalies-hypotonia-seizures syndrome 2
NM_002641.3(PIGA):c.278C>T (p.Pro93Leu)	PIGA	X	15331653	Multiple congenital anomalies-hypotonia-seizures syndrome 2
NM_002641.3(PIGA):c.355C>T (p.Arg119Trp)	PIGA	X	15331576	Multiple congenital anomalies-hypotonia-seizures syndrome 2;not provided
NM_015937.5(PIGT):c.1342C>T (p.Arg448Trp)	PIGT	20	45424323	Multiple congenital anomalies-hypotonia-seizures syndrome 3
NM_000127.2(EXT1):c.1016G>A (p.Gly339Asp)	EXT1	8	117837148	Multiple congenital exostosis
NM_000127.2(EXT1):c.1018C>T (p.Arg340Cys)	EXT1	8	117837146	Multiple congenital exostosis;not provided
NM_000459.4(TEK):c.2545C>T (p.Arg849Trp)	TEK	9	27206762	Multiple Cutaneous and Mucosal Venous Malformations
NM_000459.4(TEK):c.2744G>A (p.Arg915His)	TEK	9	27212764	Multiple Cutaneous and Mucosal Venous Malformations
NM_000244.3(MEN1):c.608G>A (p.Trp203Ter)	MEN1	11	64807952	Multiple endocrine neoplasia, type 1
NM_000244.3(MEN1):c.1322G>A (p.Trp441Ter)	MEN1	11	64805077	Multiple endocrine neoplasia, type 1
NM_130799.2(MEN1):c.778C>T (p.Gln260Ter)	MEN1	11	64807557	Multiple endocrine neoplasia, type 1
NM_000244.3(MEN1):c.778G>A (p.Glu260Lys)	MEN1	11	64807572	Multiple endocrine neoplasia, type 1
NM_000244.3(MEN1):c.1267G>A (p.Asp423Asn)	MEN1	11	64805132	Multiple endocrine neoplasia, type 1
NM_130799.2(MEN1):c.3G>A (p.Met1Ile)	MEN1	11	64810107	Multiple endocrine neoplasia, type 1
NM_130799.2(MEN1):c.85C>T (p.Arg29Ter)	MEN1	11	64810025	Multiple endocrine neoplasia, type 1
NM_130799.2(MEN1):c.1664G>A (p.Ser555Asn)	MEN1	11	64804503	Multiple endocrine neoplasia, type 1
NM_130799.2(MEN1):c.1412G>A (p.Trp471Ter)	MEN1	11	64804755	Multiple endocrine neoplasia, type 1
NM_130799.2(MEN1):c.1579C>T (p.Arg527Ter)	MEN1	11	64804588	Multiple endocrine neoplasia, type 1;not provided
NM_130799.2(MEN1):c.784-9G>A	MEN1	11	64807228	Multiple endocrine neoplasia, type 1;not provided
NM_130799.2(MEN1):c.1213C>T (p.Gln405Ter)	MEN1	11	64805171	Multiple endocrine neoplasia, type 1;not provided
NM_130799.2(MEN1):c.1243C>T (p.Arg415Ter)	MEN1	11	64805141	Multiple endocrine neoplasia, type 1;not provided
NM_020975.4(RET):c.2689C>T (p.Arg897Ter)	RET	10	43120162	Multiple endocrine neoplasia, type 2
NM_004064.4(CDKN1B):c.227G>A (p.Trp76Ter)	CDKN1B	12	12718066	Multiple endocrine neoplasia, type 4
NM_004064.4(CDKN1B):c.206C>T (p.Pro69Leu)	CDKN1B	12	12718045	Multiple endocrine neoplasia, type 4
NM_020975.4(RET):c.1826G>A (p.Cys609Tyr)	RET	10	43113622	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIA, WITH HIRSCHSPRUNG DISEASE;Medullary thyroid carcinoma;Multiple endocrine neoplasia, type 1;Multiple endocrine neoplasia, type 2;Multiple endocrine neoplasia, type 2a;Multiple endocrine neoplasia, type 2b;Multiple endocrine neoplasia, type 4;not provided
NM_000095.2(COMP):c.2152C>T (p.Arg718Trp)	COMP	19	18783129	Multiple epiphyseal dysplasia 1
NM_000095.2(COMP):c.1813G>A (p.Asp605Asn)	COMP	19	18784997	Multiple epiphyseal dysplasia 1
NM_000095.2(COMP):c.1754C>T (p.Thr585Met)	COMP	19	18785056	Multiple epiphyseal dysplasia 1;Pseudoachondroplastic spondyloepiphyseal dysplasia syndrome
NM_002381.4(MATN3):c.361C>T (p.Arg121Trp)	MATN3	2	20006173	Multiple epiphyseal dysplasia 5
NM_002381.4(MATN3):c.359C>T (p.Thr120Met)	MATN3	2	20006175	Multiple epiphyseal dysplasia 5
NM_002381.4(MATN3):c.209G>A (p.Arg70His)	MATN3	2	20012423	Multiple Epiphyseal Dysplasia, Dominant;Multiple epiphyseal dysplasia 5

NM_000401.3(EXT2):c.1404+1G>A	EXT2	11	44171743	Multiple exostoses type 2
NM_000401.3(EXT2):c.778G>A (p.Asp260Asn)	EXT2	11	44114237	Multiple exostoses type 2
NM_000401.3(EXT2):c.871C>T (p.Gln291Ter)	EXT2	11	44124817	Multiple exostoses type 2
NM_207122.1(EXT2):c.514C>T (p.Gln172Ter)	EXT2	11	44108226	Multiple exostoses type 2;not provided
NM_144997.5(FLCN):c.1532G>A (p.Trp511Ter)	FLCN	17	17214991	Multiple fibrofolliculomas
NM_144997.5(FLCN):c.1300+1G>A	FLCN	17	17216379	Multiple fibrofolliculomas
NM_144997.5(FLCN):c.1098G>A (p.Trp366Ter)	FLCN	17	17217147	Multiple fibrofolliculomas
NM_144997.5(FLCN):c.853C>T (p.Gln285Ter)	FLCN	17	17221555	Multiple fibrofolliculomas
NM_144997.5(FLCN):c.752G>A (p.Trp251Ter)	FLCN	17	17222528	Multiple fibrofolliculomas
NM_144997.5(FLCN):c.634C>T (p.Gln212Ter)	FLCN	17	17222646	Multiple fibrofolliculomas
NM_144997.5(FLCN):c.1533G>A (p.Trp511Ter)	FLCN	17	17214990	Multiple fibrofolliculomas;not provided
NM_144997.5(FLCN):c.346C>T (p.Gln116Ter)	FLCN	17	17226226	Multiple fibrofolliculomas;not provided
NM_144997.5(FLCN):c.1579C>T (p.Arg527Ter)	FLCN	17	17213816	Multiple fibrofolliculomas;not provided
NM_144997.5(FLCN):c.1432+1G>A	FLCN	17	17215184	Multiple fibrofolliculomas;not provided
NM_144997.5(FLCN):c.1429C>T (p.Arg477Ter)	FLCN	17	17215188	Multiple fibrofolliculomas;not provided
NM_001288953.1(TTC7A):c.727C>T (p.Gln243Ter)	TTC7A	2	46993514	Multiple gastrointestinal atresias
NM_001288953.1(TTC7A):c.1474C>T (p.Gln492Ter)	TTC7A	2	47024294	Multiple gastrointestinal atresias
NM_001288953.1(TTC7A):c.2392G>A (p.Ala798Thr)	TTC7A	2	47073840	Multiple gastrointestinal atresias
NM_001288953.1(TTC7A):c.109G>A (p.Glu37Lys)	TTC7A	2	46950389	Multiple gastrointestinal atresias
NM_020458.3(TTC7A):c.2470C>T (p.Gln824Ter)	TTC7A	2	47073816	Multiple gastrointestinal atresias
NM_012200.3(B3GAT3):c.830G>A (p.Arg277Gln)	B3GAT3	11	62616585	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects
NM_212552.2(BOLA3):c.136C>T (p.Arg46Ter)	BOLA3	2	74145222	Multiple mitochondrial dysfunctions syndrome 2
NM_000089.3(COL1A2):c.2549G>A (p.Gly850Glu)	COL1A2	7	94423102	Multiple prenatal fractures;Skeletal dysplasia
NM_005199.4(CHRNG):c.13C>T (p.Gln5Ter)	CHRNG	2	232539760	Multiple pterygium syndrome Escobar type
NM_005199.4(CHRNG):c.1408C>T (p.Arg470Ter)	CHRNG	2	232545570	Multiple pterygium syndrome Escobar type
NM_005199.4(CHRNG):c.136C>T (p.Arg46Ter)	CHRNG	2	232540072	Multiple pterygium syndrome Escobar type
NM_005693.3(NR1H3):c.1244G>A (p.Arg415Gln)	NR1H3	11	47268596	Multiple sclerosis
NM_004612.3(TGFBR1):c.1240C>T (p.Arg414Ter)	TGFBR1	9	99146594	Multiple self healing squamous epithelioma;Thoracic aortic aneurysm and aortic dissection
NM_182760.3(SUMF1):c.979C>T (p.Arg327Ter)	SUMF1	3	4376365	Multiple sulfatase deficiency
NM_182760.3(SUMF1):c.1045C>T (p.Arg349Trp)	SUMF1	3	4362224	Multiple sulfatase deficiency
NM_182760.3(SUMF1):c.1046G>A (p.Arg349Gln)	SUMF1	3	4362223	Multiple sulfatase deficiency
NM_182760.3(SUMF1):c.1033C>T (p.Arg345Cys)	SUMF1	3	4362236	Multiple sulfatase deficiency
NM_182760.3(SUMF1):c.653G>A (p.Cys218Tyr)	SUMF1	3	4418082	Multiple sulfatase deficiency
NM_001164675.1(SUMF1):c.836C>T (p.Ala279Val)	SUMF1	3	4417132	Multiple sulfatase deficiency;not provided
NM_000557.4(GDF5):c.1424G>A (p.Ser475Asn)	GDF5	20	35433991	Multiple synostoses syndrome 2
NM_002010.2(FGF9):c.296G>A (p.Ser99Asn)	FGF9	13	21681060	Multiple synostoses syndrome 3
m.4284G>A	MT-TI	MT	4284	Multisystem disorder;Myoclonus with epilepsy with ragged red fibers
NM_000036.2(AMPD1):c.1261C>T (p.Arg421Trp)	AMPD1	1	114677972	Muscle AMP deaminase deficiency
NM_017739.3(POMGNT1):c.1649G>A (p.Ser550Asn)	POMGNT1	1	46190473	Muscle eye brain disease
NM_001243766.1(POMGNT1):c.932G>A (p.Arg311Gln)	POMGNT1	1	46193873	Muscle eye brain disease
NM_017739.3(POMGNT1):c.1324C>T (p.Arg442Cys)	POMGNT1	1	46192397	Muscle eye brain disease;not provided
NM_017739.3(POMGNT1):c.931C>T (p.Arg311Ter)	POMGNT1	1	46193874	Muscle eye brain disease;not provided
NM_017739.3(POMGNT1):c.1895+1G>A	POMGNT1	1	46189457	Muscle eye brain disease;RETINITIS PIGMENTOSA 76
NM_005259.2(MSTN):c.373+5G>A	MSTN	2	190062219	Muscle hypertrophy
NM_016213.4(TRIP4):c.950G>A (p.Arg317Gln)	TRIP4	15	64409735	Muscular dystrophy, congenital, davignon-chauveau type

NM_005198.4(CHKB):c.922C>T (p.Gln308Ter)	CHKB	22	50579979	Muscular dystrophy, congenital, megaconial type
NM_005198.4(CHKB):c.677+1G>A	CHKB	22	50580564	Muscular dystrophy, congenital, megaconial type
NM_016532.3(INPP5K):c.67G>A (p.Val23Met)	INPP5K	17	1513957	MUSCULAR DYSTROPHY, CONGENITAL, WITH CATARACTS AND INTELLECTUAL DISABILITY
NM_016532.3(INPP5K):c.805G>A (p.Asp269Asn)	INPP5K	17	1498094	MUSCULAR DYSTROPHY, CONGENITAL, WITH CATARACTS AND INTELLECTUAL DISABILITY
NM_016532.3(INPP5K):c.418G>A (p.Gly140Ser)	INPP5K	17	1509314	MUSCULAR DYSTROPHY, CONGENITAL, WITH CATARACTS AND INTELLECTUAL DISABILITY
NM_001199563.1(BVES):c.602C>T (p.Ser201Phe)	BVES	6	105124593	Muscular dystrophy, limb-girdle, type 2X
NM_007171.3(POMT1):c.226G>A (p.Gly76Arg)	POMT1	9	131506217	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 1
NM_007171.3(POMT1):c.907C>T (p.Gln303Ter)	POMT1	9	131510401	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 1
NM_007171.3(POMT1):c.1958C>T (p.Pro653Leu)	POMT1	9	131522113	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 1
NM_007171.3(POMT1):c.1153C>T (p.Gln385Ter)	POMT1	9	131513243	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 1;not provided
NM_007171.3(POMT1):c.793C>T (p.Arg265Ter)	POMT1	9	131510287	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 1;not provided
NM_014254.2(TMEM5):c.1018C>T (p.Arg340Ter)	TMEM5	12	63808778	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 10
NM_152490.4(B3GALNT2):c.740G>A (p.Gly247Glu)	B3GALNT2	1	235470872	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 11;not provided
NM_152490.4(B3GALNT2):c.802G>A (p.Val268Met)	B3GALNT2	1	235465675	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 11;not provided
NM_152490.4(B3GALNT2):c.1423C>T (p.Gln475Ter)	B3GALNT2	1	235450286	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 11;not provided
NM_032237.4(POMK):c.325C>T (p.Gln109Ter)	POMK	8	43122149	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 12;Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 12
NM_013334.3(GMPPB):c.220C>T (p.Arg74Ter)	GMPPB	3	49723293	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 14
NM_021971.2(GMPPB):c.1000G>A (p.Asp334Asn)	GMPPB	3	49721835	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 14;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type b, 14;Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14
NM_021971.2(GMPPB):c.553C>T (p.Arg185Cys)	GMPPB	3	49722604	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 14;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type b, 14;Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14
NM_032806.5(POMGNT2):c.1333C>T (p.Arg445Ter)	POMGNT2	3	43080099	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 8
NM_032806.5(POMGNT2):c.473G>A (p.Arg158His)	POMGNT2	3	43080959	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 8;Neonatal death;Severe hydrocephalus
NM_013334.3(GMPPB):c.95C>T (p.Pro32Leu)	GMPPB	3	49723632	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type b, 14;Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14
NM_013334.3(GMPPB):c.860G>A (p.Arg287Gln)	GMPPB	3	49722056	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type b, 14;Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14
NM_013334.3(GMPPB):c.64C>T (p.Pro22Ser)	GMPPB	3	49723663	Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14
NM_013334.3(GMPPB):c.859C>T (p.Arg287Trp)	GMPPB	3	49722057	Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14
NM_013334.3(GMPPB):c.308C>T (p.Pro103Leu)	GMPPB	3	49723066	Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14
NM_013334.3(GMPPB):c.1069G>A (p.Val357Ile)	GMPPB	3	49721847	Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14;not provided
NM_013334.3(GMPPB):c.760G>A (p.Val254Met)	GMPPB	3	49722239	Muscular dystrophy-dystroglycanopathy (limb-girdle), type c, 14;not provided
NM_173660.4(DOK7):c.601C>T (p.Arg201Ter)	DOK7	4	3485607	Myasthenia, limb-girdle, familial
NM_005055.4(RAPSN):c.133G>A (p.Val45Met)	RAPSN	11	47448832	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency
NM_005055.4(RAPSN):c.484G>A (p.Glu162Lys)	RAPSN	11	47447859	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency
NM_005055.4(RAPSN):c.490C>T (p.Arg164Cys)	RAPSN	11	47447853	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency;Myopathy

NM_144988.3(ALG14):c.194C>T (p.Pro65Leu)	ALG14	1	95064960	Myasthenic syndrome, congenital, 15
NM_144988.3(ALG14):c.310C>T (p.Arg104Ter)	ALG14	1	95027239	Myasthenic syndrome, congenital, 15
NM_002334.3(LRP4):c.3697G>A (p.Glu1233Lys)	LRP4	11	46875806	Myasthenic syndrome, congenital, 17
NM_002334.3(LRP4):c.3830G>A (p.Arg1277His)	LRP4	11	46875551	Myasthenic syndrome, congenital, 17
NM_021815.4(SLC5A7):c.194G>A (p.Gly65Glu)	SLC5A7	2	107992121	Myasthenic syndrome, congenital, 20, presynaptic
NM_021815.4(SLC5A7):c.313C>T (p.Pro105Ser)	SLC5A7	2	107992992	Myasthenic syndrome, congenital, 20, presynaptic
NM_021815.4(SLC5A7):c.1082G>A (p.Arg361Gln)	SLC5A7	2	108008651	Myasthenic syndrome, congenital, 20, presynaptic
NM_000747.2(CHRNB1):c.865G>A (p.Val289Met)	CHRNB1	17	7454341	Myasthenic syndrome, congenital, 2a, slow-channel
NM_000751.2(CHRND):c.866C>T (p.Ser289Phe)	CHRND	2	232531397	Myasthenic syndrome, congenital, 3a, slow-channel
NM_000751.2(CHRND):c.238G>A (p.Glu80Lys)	CHRND	2	232527440	Myasthenic syndrome, congenital, 3b, fast-channel
NM_000751.2(CHRND):c.1204G>A (p.Glu402Lys)	CHRND	2	232534087	Myasthenic syndrome, congenital, 3c, associated with acetylcholine receptor deficiency
NM_000080.3(CHRNE):c.865C>T (p.Leu289Phe)	CHRNE	17	4900845	Myasthenic syndrome, congenital, 4a, slow-channel
NM_000080.3(CHRNE):c.721C>T (p.Leu241Phe)	CHRNE	17	4901071	Myasthenic syndrome, congenital, 4a, slow-channel
NM_000080.3(CHRNE):c.422C>T (p.Pro141Leu)	CHRNE	17	4902010	Myasthenic syndrome, congenital, 4b, fast-channel
NM_000080.3(CHRNE):c.37G>A (p.Gly13Arg)	CHRNE	17	4903027	Myasthenic syndrome, congenital, 4b, fast-channel
NM_177402.4(SYT2):c.923C>T (p.Pro308Leu)	SYT2	1	202599348	Myasthenic syndrome, congenital, 7, presynaptic
NM_005592.3(MUSK):c.2180C>T (p.Ala727Val)	MUSK	9	110800558	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency
NM_000080.3(CHRNE):c.250C>T (p.Arg84Ter)	CHRNE	17	4902311	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency
NM_000080.3(CHRNE):c.344+1G>A	CHRNE	17	4902216	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency
NM_000080.3(CHRNE):c.-96C>T	CHRNE	17	4903159	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency
NM_000080.3(CHRNE):c.991C>T (p.Arg331Trp)	CHRNE	17	4899509	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency
NM_000080.3(CHRNE):c.501-16G>A	CHRNE	17	4901641	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency
NM_001039523.2(CHRNA1):c.601G>A (p.Val201Met)	CHRNA1	2	174754233	Myasthenic syndrome, slow-channel congenital
NM_001039523.2(CHRNA1):c.896C>T (p.Thr299Ile)	CHRNA1	2	174750127	Myasthenic syndrome, slow-channel congenital
NM_001039523.2(CHRNA1):c.592G>A (p.Gly198Ser)	CHRNA1	2	174754242	Myasthenic syndrome, slow-channel congenital
NM_000079.3(CHRNA1):c.737C>T (p.Ser246Phe)	CHRNA1	2	174753544	Myasthenic syndrome, slow-channel congenital
NM_003264.4(TLR2):c.2258G>A (p.Arg753Gln)	TLR2	4	153705165	Mycobacterium tuberculosis, susceptibility to
NM_001172567.1(MYD88):c.625C>T (p.Arg209Cys)	MYD88	3	38140510	Myd88 deficiency
m.3242G>A	MT-TL1	MT	3242	Myelodysplastic syndrome
NM_003467.2(CXCR4):c.1000C>T (p.Arg334Ter)	CXCR4	2	136114928	Myelokathexis, isolated;Warts, hypogammaglobulinemia, infections, and myelokathexis
NM_000250.1(MPO):c.995C>T (p.Ala332Val)	MPO	17	58278036	Myeloperoxidase deficiency
NM_000250.1(MPO):c.1501G>A (p.Gly501Ser)	MPO	17	58273534	Myeloperoxidase deficiency
NM_000250.1(MPO):c.1495C>T (p.Arg499Cys)	MPO	17	58273540	Myeloperoxidase deficiency
NM_000250.1(MPO):c.1705C>T (p.Arg569Trp)	MPO	17	58272835	Myeloperoxidase deficiency;not provided
NM_016222.3(DDX41):c.1574G>A (p.Arg525His)	DDX41	5	177512369	Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to
NM_016222.3(DDX41):c.3G>A (p.Met1Ile)	DDX41	5	177516943	Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to
NM_016222.3(DDX41):c.490C>T (p.Arg164Trp)	DDX41	5	177515766	Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to
NM_002473.5(MYH9):c.3493C>T (p.Arg1165Cys)	MYH9	22	36295069	MYH9 related disorders;Sebastian syndrome
NM_001128425.1(MUTYH):c.1047G>A (p.Trp349Ter)	MUTYH	1	45331800	MYH-associated polyposis
NM_001128425.1(MUTYH):c.1240C>T (p.Gln414Ter)	MUTYH	1	45331503	MYH-associated polyposis
NM_001128425.1(MUTYH):c.1281G>A (p.Trp427Ter)	MUTYH	1	45331462	MYH-associated polyposis
NM_005359.5(SMAD4):c.1486C>T (p.Arg496Cys)	SMAD4	18	51078294	Myhre syndrome;not provided
NM_004631.4(LRP8):c.2855G>A (p.Arg952Gln)	LRP8	1	53247055	Myocardial infarction 1
NM_001166284.1(CCT7):c.1313C>T (p.Ser438Leu)	CCT7	2	73252803	Myocardial infarction 1
NM_003919.2(SGCE):c.289C>T (p.Arg97Ter)	SGCE	7	94628303	Myoclonic dystonia

NM_003919.2(SGCE):c.304C>T (p.Arg102Ter)	SGCE	7	94628288	Myoclonic dystonia
NM_003919.2(SGCE):c.1114C>T (p.Arg372Ter)	SGCE	7	94598914	Myoclonic dystonia
NM_001199107.1(TBC1D24):c.1544C>T (p.Ala515Val)	TBC1D24	16	2500822	Myoclonic epilepsy, familial infantile
NM_003042.3(SLC6A1):c.131G>A (p.Arg44Gln)	SLC6A1	3	11017342	Myoclonic-atonic epilepsy
NM_003042.3(SLC6A1):c.889G>A (p.Gly297Arg)	SLC6A1	3	11025812	Myoclonic-atonic epilepsy
NM_003042.3(SLC6A1):c.863C>T (p.Ala288Val)	SLC6A1	3	11025786	Myoclonic-atonic epilepsy;not provided
m.611G>A	MT-TF	MT	611	Myoclonus with epilepsy with ragged red fibers
m.8361G>A	MT-TK	MT	8361	Myoclonus with epilepsy with ragged red fibers
NM_001080114.1(LDB3):c.728C>T (p.Pro243Leu)	LDB3	10	86692544	Myofibrillar myopathy
NM_001927.3(DES):c.735G>A (p.Glu245=)	DES	2	219420346	Myofibrillar myopathy 1
NM_001927.3(DES):c.394C>T (p.Gln132Ter)	DES	2	219418856	Myofibrillar myopathy 1
NM_001927.3(DES):c.1216C>T (p.Arg406Trp)	DES	2	219421532	Myofibrillar myopathy 1;not provided
NM_001927.3(DES):c.1325C>T (p.Thr442Ile)	DES	2	219425699	Myofibrillar myopathy 1;not provided
NM_001927.3(DES):c.1255C>T (p.Pro419Ser)	DES	2	219423787	Myofibrillar myopathy 1;not provided
NM_001927.3(DES):c.38C>T (p.Ser13Phe)	DES	2	219418500	Myofibrillar myopathy 1;Primary dilated cardiomyopathy;not provided
NM_004281.3(BAG3):c.626C>T (p.Pro209Leu)	BAG3	10	119672373	Myofibrillar myopathy, BAG3-related;not provided
NM_001458.4(FLNC):c.8130G>A (p.Trp2710Ter)	FLNC	7	128858475	Myofibrillar myopathy, filamin C-related
NM_001080116.1(LDB3):c.440C>T (p.Ala147Val)	LDB3	10	86687164	Myofibrillar myopathy, ZASP-related
NM_007078.2(LDB3):c.690-4733G>A	LDB3	10	86687163	Myofibrillar myopathy, ZASP-related;not provided
NM_001927.3(DES):c.1285C>T (p.Arg429Ter)	DES	2	219423817	Myofibrillar myopathy;Myofibrillar myopathy 1
NM_001080114.1(LDB3):c.494C>T (p.Ala165Val)	LDB3	10	86687218	Myofibrillar myopathy;Myofibrillar myopathy, ZASP-related;Primary dilated cardiomyopathy
NM_001927.3(DES):c.46C>T (p.Arg16Cys)	DES	2	219418508	Myofibrillar myopathy;not provided
NM_145693.3(LPIN1):c.1162C>T (p.Arg388Ter)	LPIN1	2	11783834	Myoglobinuria, acute recurrent, autosomal recessive
NM_145693.3(LPIN1):c.2401C>T (p.Arg801Ter)	LPIN1	2	11819590	Myoglobinuria, acute recurrent, autosomal recessive
m.5920G>A	MT-CO1	MT	5920	Myoglobinuria, recurrent
m.15990C>T	MT-TP	MT	15990	Myopathy
NM_133378.4(TTN):c.25513C>T (p.Gln8505Ter)	TTN	2	178706629	Myopathy
NM_004369.3(COL6A3):c.6181C>T (p.Arg2061Ter)	COL6A3	2	237361150	Myopathy
NM_000540.2(RYR1):c.3381+1G>A	RYR1	19	38467813	Myopathy
NM_004369.3(COL6A3):c.9193C>T (p.Gln3065Ter)	COL6A3	2	237334662	Myopathy
NM_001849.3(COL6A2):c.1458+1G>A	COL6A2	21	46121124	Myopathy
NM_001267550.2(TTN):c.74567G>A (p.Trp24856Ter)	TTN	2	178571565	Myopathy
NM_006077.3(MICU1):c.741+1G>A	MICU1	10	72477173	Myopathy with extrapyramidal signs
NM_213595.3(ISCU):c.149G>A (p.Gly50Glu)	ISCU	12	108564313	Myopathy with lactic acidosis, hereditary
NM_001159702.2(FHL1):c.838G>A (p.Val280Met)	FHL1	X	136209392	Myopathy with postural muscle atrophy, X-linked
NM_001449.4(FHL1):c.688+1G>A	FHL1	X	136208642	Myopathy with postural muscle atrophy, X-linked
NM_032446.2(MEGF10):c.1559G>A (p.Trp520Ter)	MEGF10	5	127420176	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset
NM_032446.2(MEGF10):c.211C>T (p.Arg71Trp)	MEGF10	5	127339214	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant
NM_005876.4(SPEG):c.6697C>T (p.Gln2233Ter)	SPEG	2	219484160	Myopathy, centronuclear, 5
NM_005876.4(SPEG):c.4276C>T (p.Arg1426Ter)	SPEG	2	219473732	Myopathy, centronuclear, 5
NM_001005360.2(DNM2):c.1106G>A (p.Arg369Gln)	DNM2	19	10793833	Myopathy, centronuclear;Myopathy, centronuclear, 1
NM_001005360.2(DNM2):c.1105C>T (p.Arg369Trp)	DNM2	19	10793832	Myopathy, centronuclear;Myopathy, centronuclear, 1
NM_001005360.2(DNM2):c.1102G>A (p.Glu368Lys)	DNM2	19	10793829	Myopathy, centronuclear;Myopathy, centronuclear, 1
NM_001005360.2(DNM2):c.1393C>T (p.Arg465Trp)	DNM2	19	10798543	Myopathy, centronuclear;Myopathy, centronuclear, 1;not provided
NM_001005360.2(DNM2):c.1565G>A (p.Arg522His)	DNM2	19	10812271	Myopathy, centronuclear;not provided

NM_001005360.2(DNM2):c.1856C>T (p.Ser619Leu)	DNM2	19	10823862	Myopathy, centronuclear;Severe X-linked myotubular myopathy
NM_005343.3(HRAS):c.187G>A (p.Glu63Lys)	HRAS	11	533869	Myopathy, congenital, with excess of muscle spindles;not provided
NM_000257.3(MYH7):c.5566G>A (p.Glu1856Lys)	MYH7	14	23414096	Myopathy, distal, 1
NM_199189.2(MATR3):c.460C>T (p.Pro154Ser)	MATR3	5	139307875	Myopathy, distal, 2
NM_001458.4(FLNC):c.577G>A (p.Ala193Thr)	FLNC	7	128835550	Myopathy, distal, 4;not provided
NM_152328.4(ADSSL1):c.781G>A (p.Asp261Asn)	ADSSL1	14	104741231	Myopathy, distal, 5
NM_001002020.2(PUS1):c.346C>T (p.Arg116Trp)	PUS1	12	131932301	Myopathy, lactic acidosis, and sideroblastic anemia 1
NM_001002020.2(PUS1):c.799C>T (p.Arg267Trp)	PUS1	12	131941630	Myopathy, lactic acidosis, and sideroblastic anemia 1
NM_001040436.2(YARS2):c.572G>A (p.Gly191Asp)	YARS2	12	32755303	Myopathy, lactic acidosis, and sideroblastic anemia 2
NM_001040436.2(YARS2):c.1078C>T (p.Arg360Ter)	YARS2	12	32750744	Myopathy, lactic acidosis, and sideroblastic anemia 2
NM_001040436.2(YARS2):c.137G>A (p.Gly46Asp)	YARS2	12	32755738	Myopathy, lactic acidosis, and sideroblastic anemia 2;not provided
NC_012920.1:m.8969G>A	MT-ATP6	MT	8969	Myopathy, lactic acidosis, and sideroblastic anemia 3
NM_005262.2(GFER):c.581G>A (p.Arg194His)	GFER	16	1985991	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay;not provided
m.622G>A	MT-TF	MT	622	Myopathy, mitochondrial, late-onset
NM_024854.3(PYROXD1):c.285+1G>A	PYROXD1	12	21445467	MYOPATHY, MYOFIBRILLAR, 8
NM_024854.3(PYROXD1):c.414+1G>A	PYROXD1	12	21449692	MYOPATHY, MYOFIBRILLAR, 8
NM_001159702.2(FHL1):c.458G>A (p.Cys153Tyr)	FHL1	X	136207918	Myopathy, reducing body, X-linked, childhood-onset
NM_001159702.2(FHL1):c.367C>T (p.His123Tyr)	FHL1	X	136207827	Myopathy, reducing body, X-linked, early-onset, severe
NM_001159702.2(FHL1):c.449G>A (p.Cys150Tyr)	FHL1	X	136207909	Myopathy, reducing body, X-linked, early-onset, severe
NM_032790.3(ORAI1):c.734C>T (p.Pro245Leu)	ORAI1	12	121641471	Myopathy, tubular aggregate, 2
NM_032790.3(ORAI1):c.292G>A (p.Gly98Ser)	ORAI1	12	121627039	Myopathy, tubular aggregate, 2
NM_032790.3(ORAI1):c.412C>T (p.Leu138Phe)	ORAI1	12	121641149	Myopathy, tubular aggregate, 2
NM_000018.3(ACADVL):c.1376G>A (p.Arg459Gln)	ACADVL	17	7224011	Myopathy;Rhabdomyolysis;Very long chain acyl-CoA dehydrogenase deficiency;not provided
NM_001099679.1(TRIM32):c.1459G>A (p.Asp487Asn)	TRIM32	9	116699201	Myopathy;Sarcotubular myopathy
NM_201269.2(ZNF644):c.*592G>A	ZNF644	1	90916206	Myopia 21, autosomal dominant
NM_001169109.1(SCO2):c.341G>A (p.Arg114His)	SCO2	22	50524071	Myopia 6
NM_001134418.1(P3H2):c.13C>T (p.Gln5Ter)	P3H2	3	189995367	Myopia, high, with cataract and vitreoretinal degeneration
NM_001849.3(COL6A2):c.2455C>T (p.Gln819Ter)	COL6A2	21	46126535	Myosclerosis, autosomal recessive;not provided
NM_000257.3(MYH7):c.5533C>T (p.Arg1845Trp)	MYH7	14	23415021	Myosin storage myopathy;Scapuloperoneal myopathy, MYH7-related
NM_006790.2(MYOT):c.179C>T (p.Ser60Phe)	MYOT	5	137870830	Myotilinopathy
NM_000083.2(CLCN1):c.2635C>T (p.Gln879Ter)	CLCN1	7	143351633	Myotonia
NM_000083.2(CLCN1):c.1013G>A (p.Arg338Gln)	CLCN1	7	143331265	Myotonia congenita
NM_000083.2(CLCN1):c.1412C>T (p.Ser471Phe)	CLCN1	7	143339263	Myotonia congenita
NM_000083.2(CLCN1):c.1592C>T (p.Ala531Val)	CLCN1	7	143341938	Myotonia congenita
NM_000083.2(CLCN1):c.577G>A (p.Glu193Lys)	CLCN1	7	143321729	Myotonia congenita
NM_000083.2(CLCN1):c.803C>T (p.Thr268Met)	CLCN1	7	143324442	Myotonia congenita
NM_000083.2(CLCN1):c.847C>T (p.Leu283Phe)	CLCN1	7	143324486	Myotonia congenita
NM_000083.2(CLCN1):c.929C>T (p.Thr310Met)	CLCN1	7	143330847	Myotonia congenita
NM_000083.2(CLCN1):c.1649C>T (p.Thr550Met)	CLCN1	7	143341995	Myotonia congenita
NM_000083.2(CLCN1):c.937G>A (p.Ala313Thr)	CLCN1	7	143330855	Myotonia congenita;not provided
NM_000083.2(CLCN1):c.774+1G>A	CLCN1	7	143323387	Myotonia congenita;not provided
m.5650G>A	MT-TA	MT	5650	Myotonic dystrophy-like myopathy
NM_005850.4(SF3B4):c.913+1G>A	SF3B4	1	149925835	Nager syndrome
NM_005850.4(SF3B4):c.1006C>T (p.Arg336Ter)	SF3B4	1	149923922	Nager syndrome

NM_005850.4(SF3B4):c.625C>T (p.Gln209Ter)	SF3B4	1	149926457	Nager syndrome
NM_003506.3(FZD6):c.1531C>T (p.Arg511Cys)	FZD6	8	103328406	Nail disease;Nail disorder, nonsyndromic congenital, 10
NM_002316.3(LMX1B):c.661C>T (p.Arg221Ter)	LMX1B	9	126693243	Nail-patella syndrome
NM_002316.3(LMX1B):c.244C>T (p.Gln82Ter)	LMX1B	9	126615487	Nail-patella syndrome
NM_002316.3(LMX1B):c.668G>A (p.Arg223Gln)	LMX1B	9	126693250	Nail-patella syndrome
NM_002316.3(LMX1B):c.745C>T (p.Arg249Ter)	LMX1B	9	126693527	Nail-patella syndrome
NM_002316.3(LMX1B):c.691C>T (p.Arg231Ter)	LMX1B	9	126693273	Nail-patella syndrome;not provided
NM_198270.3(NHS):c.115C>T (p.Gln39Ter)	NHS	X	17375872	Nance-Horan syndrome
NM_198270.3(NHS):c.1117C>T (p.Arg373Ter)	NHS	X	17724370	Nance-Horan syndrome;not provided
NM_002447.3(MST1R):c.917G>A (p.Arg306His)	MST1R	3	49902693	Nasopharyngeal carcinoma, susceptibility to, 3
NM_002437.4(MPV17):c.149G>A (p.Arg50Gln)	MPV17	2	27313031	Navajo neurohepatopathy
NM_002437.4(MPV17):c.359G>A (p.Trp120Ter)	MPV17	2	27312510	Navajo neurohepatopathy
NM_002437.4(MPV17):c.206G>A (p.Trp69Ter)	MPV17	2	27312753	Navajo neurohepatopathy
NM_002437.4(MPV17):c.509C>T (p.Ser170Phe)	MPV17	2	27309934	Navajo neurohepatopathy
NM_002437.4(MPV17):c.70+5G>A	MPV17	2	27322443	Navajo neurohepatopathy
NM_002437.4(MPV17):c.106C>T (p.Gln36Ter)	MPV17	2	27313074	Navajo neurohepatopathy
NM_002437.4(MPV17):c.148C>T (p.Arg50Trp)	MPV17	2	27313032	Navajo neurohepatopathy;not provided
NM_002230.2(JUP):c.468G>A (p.Pro156=)	JUP	17	41769418	Naxos disease
NM_002230.2(JUP):c.1615C>T (p.Gln539Ter)	JUP	17	41758753	Naxos disease
NM_002230.2(JUP):c.794G>A (p.Arg265His)	JUP	17	41767494	Naxos disease
NM_152263.3(TPM3):c.855-1G>A	TPM3	1	154167941	Nemaline myopathy 1;not provided
NM_152263.3(TPM3):c.94C>T (p.Gln32Ter)	TPM3	1	154191925	Nemaline myopathy 1;not provided
NM_198271.4(LMOD3):c.1201C>T (p.Arg401Ter)	LMOD3	3	69119154	Nemaline myopathy 10
NM_032578.3(MYPN):c.1129C>T (p.Arg377Ter)	MYPN	10	68145525	NEMALINE MYOPATHY 11, AUTOSOMAL RECESSIVE
NM_032578.3(MYPN):c.3169C>T (p.Arg1057Ter)	MYPN	10	68197362	NEMALINE MYOPATHY 11, AUTOSOMAL RECESSIVE
NM_032578.3(MYPN):c.3214C>T (p.Arg1072Ter)	MYPN	10	68197407	NEMALINE MYOPATHY 11, AUTOSOMAL RECESSIVE
NM_001271208.1(NEB):c.6385C>T (p.Gln2129Ter)	NEB	2	151656263	Nemaline myopathy 2
NM_004543.4(NEB):c.3987+1G>A	NEB	2	151674476	Nemaline myopathy 2
NM_004543.4(NEB):c.3567+1G>A	NEB	2	151677875	Nemaline myopathy 2, autosomal recessive
NM_004543.4(NEB):c.3774+1G>A	NEB	2	151677564	Nemaline myopathy 2, autosomal recessive
NM_001271208.1(NEB):c.1152+1G>A	NEB	2	151706880	Nemaline myopathy 2;not provided
NM_001100.3(ACTA1):c.493G>A (p.Val165Met)	ACTA1	1	229432393	Nemaline myopathy 3
NM_003289.3(TPM2):c.349G>A (p.Glu117Lys)	TPM2	9	35685672	Nemaline myopathy 4;not provided
NM_003289.3(TPM2):c.121G>A (p.Glu41Lys)	TPM2	9	35689265	Nemaline myopathy 4;not provided
NM_001101362.2(KBTBD13):c.1222C>T (p.Arg408Cys)	KBTBD13	15	65078037	Nemaline myopathy 6
NM_138638.4(CFL2):c.103G>A (p.Ala35Thr)	CFL2	14	34713462	Nemaline myopathy 7
NM_138638.4(CFL2):c.19G>A (p.Val7Met)	CFL2	14	34713546	Nemaline myopathy 7
NM_152393.3(KLHL40):c.1582G>A (p.Glu528Lys)	KLHL40	3	42689029	Nemaline myopathy 8
NM_152393.3(KLHL40):c.602G>A (p.Trp201Ter)	KLHL40	3	42686220	Nemaline myopathy 8
NM_006063.2(KLHL41):c.1238C>T (p.Ser413Leu)	KLHL41	2	169514701	Nemaline myopathy 9
NM_000352.4(ABCC8):c.3545G>A (p.Arg1182Gln)	ABCC8	11	17404524	Neonatal diabetes mellitus;Transient neonatal diabetes mellitus 2
NM_014251.2(SLC25A13):c.1078C>T (p.Arg360Ter)	SLC25A13	7	96184376	Neonatal intrahepatic cholestasis caused by citrin deficiency
NM_014251.2(SLC25A13):c.1592G>A (p.Gly531Asp)	SLC25A13	7	96121997	Neonatal intrahepatic cholestasis caused by citrin deficiency
NM_014251.2(SLC25A13):c.15G>A (p.Lys5=)	SLC25A13	7	96321942	Neonatal intrahepatic cholestasis caused by citrin deficiency
NM_014251.2(SLC25A13):c.1801G>A (p.Glu601Lys)	SLC25A13	7	96121695	Neonatal intrahepatic cholestasis caused by citrin deficiency

NM_014251.2(SLC25A13):c.550C>T (p.Arg184Ter)	SLC25A13	7	96193102	Neonatal intrahepatic cholestasis caused by citrin deficiency
NM_014251.2(SLC25A13):c.615+5G>A	SLC25A13	7	96193032	Neonatal intrahepatic cholestasis caused by citrin deficiency
NM_014251.2(SLC25A13):c.775C>T (p.Gln259Ter)	SLC25A13	7	96189654	Neonatal intrahepatic cholestasis caused by citrin deficiency
NM_014251.2(SLC25A13):c.1813C>T (p.Arg605Ter)	SLC25A13	7	96121683	Neonatal intrahepatic cholestasis caused by citrin deficiency;not provided
NM_000388.3(CASR):c.2009G>A (p.Gly670Glu)	CASR	3	122283963	Neonatal severe hyperparathyroidism
NM_000388.3(CASR):c.553C>T (p.Arg185Ter)	CASR	3	122261588	Neonatal severe hyperparathyroidism
NM_004304.4(ALK):c.3824G>A (p.Arg1275Gln)	ALK	2	29209798	Neoplasm of brain;Neoplasm of breast;Neuroblastoma;Neuroblastoma 3
NM_000314.6(PTEN):c.518G>A (p.Arg173His)	PTEN	10	87952143	Neoplasm of brain;not provided
NM_004448.3(ERBB2):c.2686C>T (p.Arg896Cys)	ERBB2	17	39725363	Neoplasm of breast
NM_000038.5(APC):c.3359G>A (p.Gly1120Glu)	APC	5	112838953	Neoplasm of stomach
NM_000038.5(APC):c.3199C>T (p.Gln1067Ter)	APC	5	112838793	Neoplasm of stomach
NM_001128425.1(MUTYH):c.1213C>T (p.Pro405Ser)	MUTYH	1	45331530	Neoplasm of stomach
NM_000321.2(RB1):c.1666C>T (p.Arg556Ter)	RB1	13	48381414	Neoplasm;Retinoblastoma;Retinoblastoma, trilateral
NM_000054.4(AVPR2):c.607C>T (p.Arg203Cys)	AVPR2	X	153906113	Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.337C>T (p.Arg113Trp)	AVPR2	X	153905843	Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.213G>A (p.Trp71Ter)	AVPR2	X	153905719	Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.253G>A (p.Asp85Asn)	AVPR2	X	153905759	Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.602G>A (p.Gly201Asp)	AVPR2	X	153906108	Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.410G>A (p.Arg137His)	AVPR2	X	153905916	Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.541C>T (p.Arg181Cys)	AVPR2	X	153906047	Nephrogenic diabetes insipidus, X-linked
NM_000486.5(AQP2):c.763C>T (p.Gln255Ter)	AQP2	12	49955555	Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.878G>A (p.Trp293Ter)	AVPR2	X	153906384	Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.1009C>T (p.Arg337Ter)	AVPR2	X	153906621	Nephrogenic diabetes insipidus, X-linked;not provided
NM_000054.4(AVPR2):c.310C>T (p.Arg104Cys)	AVPR2	X	153905816	Nephrogenic diabetes insipidus;Nephrogenic diabetes insipidus, X-linked
NM_000054.4(AVPR2):c.409C>T (p.Arg137Cys)	AVPR2	X	153905915	Nephrogenic syndrome of inappropriate antidiuresis
NM_001127899.3(CLCN5):c.2320C>T (p.Arg774Ter)	CLCN5	X	50090846	Nephrolithiasis, X-linked recessive
NM_001127899.3(CLCN5):c.1727G>A (p.Gly576Glu)	CLCN5	X	50088867	Nephrolithiasis, X-linked recessive
NM_003052.4(SLC34A1):c.439G>A (p.Val147Met)	SLC34A1	5	177386473	Nephrolithiasis/osteoporosis, hypophosphatemic, 1
NM_153240.4(NPHP3):c.1817G>A (p.Trp606Ter)	NPHP3	3	132699988	Nephronophthisis
NM_000272.3(NPHP1):c.1027G>A (p.Gly343Arg)	NPHP1	2	110163048	Nephronophthisis 1
NM_001142301.1(TMEM67):c.2218G>A (p.Gly740Ser)	TMEM67	8	93808861	Nephronophthisis 11
NM_024753.4(TTC21B):c.626C>T (p.Pro209Leu)	TTC21B	2	165941111	Nephronophthisis 12
NM_025132.3(WDR19):c.682C>T (p.Gln228Ter)	WDR19	4	39205232	Nephronophthisis 13
NM_025132.3(WDR19):c.3703G>A (p.Glu1235Lys)	WDR19	4	39274945	Nephronophthisis 13
NM_001271620.2(ZNF423):c.2558C>T (p.Pro853Leu)	ZNF423	16	49636414	Nephronophthisis 14
NM_014956.4(CEP164):c.277C>T (p.Arg93Trp)	CEP164	11	117351872	Nephronophthisis 15
NM_014956.4(CEP164):c.1573C>T (p.Gln525Ter)	CEP164	11	117381864	Nephronophthisis 15
NM_014956.4(CEP164):c.1726C>T (p.Arg576Ter)	CEP164	11	117387204	Nephronophthisis 15
NM_016122.2(CEP83):c.121C>T (p.Arg41Ter)	CEP83	12	94412370	Nephronophthisis 18
NM_016122.2(CEP83):c.241C>T (p.Gln81Ter)	CEP83	12	94411780	Nephronophthisis 18
NM_016122.2(CEP83):c.625C>T (p.Arg209Ter)	CEP83	12	94378967	Nephronophthisis 18
NM_001128608.1(MAPKBP1):c.592C>T (p.Arg198Ter)	MAPKBP1	15	41812609	Nephronophthisis 20
NM_001128608.1(MAPKBP1):c.4393C>T (p.Arg1465Ter)	MAPKBP1	15	41825284	Nephronophthisis 20
NM_001128608.1(MAPKBP1):c.1318C>T (p.Arg440Ter)	MAPKBP1	15	41815388	Nephronophthisis 20
NM_001128608.1(MAPKBP1):c.1631G>A (p.Arg544Gln)	MAPKBP1	15	41816937	Nephronophthisis 20

NM_001128608.1(MAPKBP1):c.2444-1G>A	MAPKBP1	15	41819594	Nephronophthisis 20
NM_001128608.1(MAPKBP1):c.2827C>T (p.Gln943Ter)	MAPKBP1	15	41821674	Nephronophthisis 20
NM_015102.4(NPHP4):c.2377C>T (p.Gln793Ter)	NPHP4	1	5887394	Nephronophthisis 4
NM_178170.2(NEK8):c.1273C>T (p.His425Tyr)	NEK8	17	28738721	Nephronophthisis 9
NM_015102.4(NPHP4):c.2335C>T (p.Gln779Ter)	NPHP4	1	5887436	Nephronophthisis;Senior-Loken syndrome 4
NM_015102.4(NPHP4):c.1972C>T (p.Arg658Ter)	NPHP4	1	5904788	Nephronophthisis;Senior-Loken syndrome 4
NM_001023570.3(IQCB1):c.1381C>T (p.Arg461Ter)	IQCB1	3	121781772	Nephronophthisis;Senior-Loken syndrome 5;not provided
NM_004937.2(CTNS):c.414G>A (p.Trp138Ter)	CTNS	17	3655305	Nephropathic cystinosis
NM_001031681.2(CTNS):c.506G>A (p.Gly169Asp)	CTNS	17	3656531	Nephropathic cystinosis
NM_004937.2(CTNS):c.1015G>A (p.Gly339Arg)	CTNS	17	3660280	Nephropathic cystinosis
NM_004937.2(CTNS):c.613G>A (p.Asp205Asn)	CTNS	17	3656727	Nephropathic cystinosis
NM_014625.3(NPHS2):c.412C>T (p.Arg138Ter)	NPHS2	1	179561328	Nephrotic syndrome, idiopathic, steroid-resistant
NM_014625.3(NPHS2):c.59C>T (p.Pro20Leu)	NPHS2	1	179575806	Nephrotic syndrome, idiopathic, steroid-resistant
NM_014625.3(NPHS2):c.538G>A (p.Val180Met)	NPHS2	1	179557227	Nephrotic syndrome, idiopathic, steroid-resistant
NM_014625.3(NPHS2):c.871C>T (p.Arg291Trp)	NPHS2	1	179552605	Nephrotic syndrome, idiopathic, steroid-resistant
NM_014625.3(NPHS2):c.868G>A (p.Val290Met)	NPHS2	1	179552608	Nephrotic syndrome, idiopathic, steroid-resistant
NM_014625.3(NPHS2):c.812C>T (p.Pro271Leu)	NPHS2	1	179552664	Nephrotic syndrome, idiopathic, steroid-resistant
NM_014625.3(NPHS2):c.115C>T (p.Gln39Ter)	NPHS2	1	179575750	Nephrotic syndrome, idiopathic, steroid-resistant
NM_014625.3(NPHS2):c.452-1G>A	NPHS2	1	179559762	Nephrotic syndrome, idiopathic, steroid-resistant
NM_014625.3(NPHS2):c.413G>A (p.Arg138Gln)	NPHS2	1	179561327	Nephrotic syndrome, idiopathic, steroid-resistant;Steroid-resistant nephrotic syndrome;not provided
NM_001424.5(EMP2):c.184C>T (p.Gln62Ter)	EMP2	16	10538060	Nephrotic syndrome, type 10
NM_001424.5(EMP2):c.28G>A (p.Ala10Thr)	EMP2	16	10547590	Nephrotic syndrome, type 10
NM_020401.3(NUP107):c.969+1G>A	NUP107	12	68713809	Nephrotic syndrome, type 11
NM_014669.4(NUP93):c.1537+1G>A	NUP93	16	56833407	Nephrotic syndrome, type 12
NM_014669.4(NUP93):c.1162C>T (p.Arg388Trp)	NUP93	16	56831918	Nephrotic syndrome, type 12
NM_016341.3(PLCE1):c.1477C>T (p.Arg493Ter)	PLCE1	10	94132444	Nephrotic syndrome, type 3
NM_016341.3(PLCE1):c.3346C>T (p.Arg1116Ter)	PLCE1	10	94254256	Nephrotic syndrome, type 3
NM_016341.3(PLCE1):c.4846C>T (p.Gln1616Ter)	PLCE1	10	94283840	Nephrotic syndrome, type 3
NM_016341.3(PLCE1):c.5560C>T (p.Gln1854Ter)	PLCE1	10	94304583	Nephrotic syndrome, type 3
NM_016341.3(PLCE1):c.4451C>T (p.Ser1484Leu)	PLCE1	10	94270547	Nephrotic syndrome, type 3
NM_016341.3(PLCE1):c.6448C>T (p.Arg2150Ter)	PLCE1	10	94322006	Nephrotic syndrome, type 3
NM_016341.3(PLCE1):c.961C>T (p.Arg321Ter)	PLCE1	10	94032007	Nephrotic syndrome, type 3
NM_016341.3(PLCE1):c.3736C>T (p.Arg1246Ter)	PLCE1	10	94259072	Nephrotic syndrome, type 3
NM_002292.3(LAMB2):c.737G>A (p.Arg246Gln)	LAMB2	3	49131128	Nephrotic syndrome, type 5, with or without ocular abnormalities
NM_003647.2(DGKE):c.127C>T (p.Gln43Ter)	DGKE	17	56834922	Nephrotic syndrome, type 7
NM_003647.2(DGKE):c.1376G>A (p.Trp459Ter)	DGKE	17	56861882	Nephrotic syndrome, type 7
NM_004309.5(ARHGDIA):c.358C>T (p.Arg120Ter)	ARHGDIA	17	81869230	Nephrotic syndrome, type 8
NM_024876.3(COQ8B):c.532C>T (p.Arg178Trp)	COQ8B	19	40705140	Nephrotic syndrome, type 9
NM_024876.3(COQ8B):c.958C>T (p.Arg320Trp)	COQ8B	19	40700387	Nephrotic syndrome, type 9
NM_024876.3(COQ8B):c.1027C>T (p.Arg343Trp)	COQ8B	19	40700318	Nephrotic syndrome, type 9
NM_024876.3(COQ8B):c.1430G>A (p.Arg477Gln)	COQ8B	19	40692240	Nephrotic syndrome, type 9
NM_024876.3(COQ8B):c.101G>A (p.Trp34Ter)	COQ8B	19	40714532	Nephrotic syndrome, type 9
NM_003860.3(BANF1):c.34G>A (p.Ala12Thr)	BANF1	11	66003284	Nestor-Guillermo progeria syndrome
NM_006846.3(SPINK5):c.2368C>T (p.Arg790Ter)	SPINK5	5	148120063	Netherton syndrome
NM_006846.3(SPINK5):c.652C>T (p.Arg218Ter)	SPINK5	5	148091214	Netherton syndrome

NM_006623.3(PHGDH):c.488G>A (p.Arg163Gln)	PHGDH	1	119727080	Neu-Laxova syndrome 1
NM_006623.3(PHGDH):c.793G>A (p.Glu265Lys)	PHGDH	1	119737114	Neu-Laxova syndrome 1
NM_006623.3(PHGDH):c.418G>A (p.Gly140Arg)	PHGDH	1	119727010	Neu-Laxova syndrome 1;not provided
NM_058179.3(PSAT1):c.536C>T (p.Ser179Leu)	PSAT1	9	78306452	Neu-laxova syndrome 2
NM_058179.3(PSAT1):c.296C>T (p.Ala99Val)	PSAT1	9	78304839	Neu-laxova syndrome 2
NM_015074.3(KIF1B):c.3649C>T (p.Pro1217Ser)	KIF1B	1	10345943	Neuroblastoma 1
NM_015074.3(KIF1B):c.4442G>A (p.Ser1481Asn)	KIF1B	1	10365476	Neuroblastoma 1;Pheochromocytoma
NM_003924.3(PHOX2B):c.590G>A (p.Gly197Asp)	PHOX2B	4	41746162	Neuroblastoma 2
NM_004304.4(ALK):c.3452C>T (p.Thr1151Met)	ALK	2	29222407	Neuroblastoma;Neuroblastoma 3
NM_153638.3(PANK2):c.894G>A (p.Met298Ile)	PANK2	20	3908191	Neurodegeneration
NM_153638.3(PANK2):c.1069C>T (p.Arg357Trp)	PANK2	20	3910664	Neurodegeneration
NM_001142298.1(SQSTM1):c.34C>T (p.Arg12Ter)	SQSTM1	5	179823038	NEURODEGENERATION WITH ATAXIA, DYSTONIA, AND GAZE PALSY, CHILDHOOD-ONSET
NM_007075.3(WDR45):c.400C>T (p.Arg134Ter)	WDR45	X	49076469	Neurodegeneration with brain iron accululation 5;Neurodegeneration with brain iron accumulation 5;not provided
NM_153638.3(PANK2):c.832C>T (p.Arg278Cys)	PANK2	20	3908129	Neurodegeneration with brain iron accumulation 1, atypical
NM_003560.3(PLA2G6):c.238G>A (p.Ala80Thr)	PLA2G6	22	38145625	Neurodegeneration with brain iron accumulation 2b
NM_001256047.1(C19orf12):c.215C>T (p.Pro72Leu)	C19orf12	19	29702923	Neurodegeneration with brain iron accumulation 4
NM_001031726.3(C19orf12):c.32C>T (p.Thr11Met)	C19orf12	19	29708415	Neurodegeneration with brain iron accumulation 4;not provided
NM_007075.3(WDR45):c.637C>T (p.Gln213Ter)	WDR45	X	49075636	Neurodegeneration with brain iron accumulation 5
NM_007075.3(WDR45):c.614G>A (p.Gly205Asp)	WDR45	X	49075659	Neurodegeneration with brain iron accumulation 5
NM_025233.6(COASY):c.1495C>T (p.Arg499Cys)	COASY	17	42565668	Neurodegeneration with brain iron accumulation 6
NM_025233.6(COASY):c.175C>T (p.Gln59Ter)	COASY	17	42562797	Neurodegeneration with brain iron accumulation 6
NM_004181.4(UCHL1):c.533G>A (p.Arg178Gln)	UCHL1	4	41264109	Neurodegeneration with optic atrophy, childhood-onset
NM_000405.4(GM2A):c.164C>T (p.Pro55Leu)	GM2A	5	151259837	Neurodegenerative illness progressing to crippling dystonia and death with relentless cerebral atrophy;Tay-Sachs disease, variant AB
NM_052876.3(NACC1):c.892C>T (p.Arg298Trp)	NACC1	19	13136099	NEURODEVELOPMENTAL DISORDER WITH EPILEPSY, CATARACTS, FEEDING DIFFICULTIES, AND DELAYED BRAIN MYELINATION
NM_020760.3(HECW2):c.3572G>A (p.Arg1191Gln)	HECW2	2	196242162	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA, SEIZURES, AND ABSENT LANGUAGE
NM_020760.3(HECW2):c.3988C>T (p.Arg1330Trp)	HECW2	2	196225800	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA, SEIZURES, AND ABSENT LANGUAGE
NM_012102.3(RERE):c.3466G>A (p.Gly1156Arg)	RERE	1	8359916	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart
NM_000146.3(FTL):c.286G>A (p.Ala96Thr)	FTL	19	48966317	Neuroferritinopathy
NM_000267.3(NF1):c.3113+1G>A	NF1	17	31230383	Neurofibromatosis, type 1
NM_000267.3(NF1):c.1260+1G>A	NF1	17	31201486	Neurofibromatosis, type 1
NM_000267.3(NF1):c.4021C>T (p.Gln1341Ter)	NF1	17	31249030	Neurofibromatosis, type 1
NM_000267.3(NF1):c.1713G>A (p.Trp571Ter)	NF1	17	31221921	Neurofibromatosis, type 1
NM_000267.3(NF1):c.6999+1G>A	NF1	17	31340646	Neurofibromatosis, type 1
NM_000267.3(NF1):c.7419G>A (p.Trp2473Ter)	NF1	17	31352281	Neurofibromatosis, type 1
NM_000267.3(NF1):c.1185+1G>A	NF1	17	31201160	Neurofibromatosis, type 1
NM_000267.3(NF1):c.5205+5G>A	NF1	17	31326257	Neurofibromatosis, type 1
NM_000267.3(NF1):c.5458C>T (p.Gln1820Ter)	NF1	17	31327751	Neurofibromatosis, type 1
NM_000267.3(NF1):c.2352G>A (p.Trp784Ter)	NF1	17	31227549	Neurofibromatosis, type 1
NM_000267.3(NF1):c.3565C>T (p.Gln1189Ter)	NF1	17	31233070	Neurofibromatosis, type 1
NM_000267.3(NF1):c.5608C>T (p.Gln1870Ter)	NF1	17	31330357	Neurofibromatosis, type 1
NM_001042492.2(NF1):c.2709G>A (p.Val903=)	NF1	17	31229324	Neurofibromatosis, type 1
NM_000267.3(NF1):c.3763C>T (p.Gln1255Ter)	NF1	17	31235665	Neurofibromatosis, type 1

NM_000267.3(NF1):c.3916C>T (p.Arg1306Ter)	NF1	17	31235963	Neurofibromatosis, type 1
NM_000267.3(NF1):c.7044G>A (p.Trp2348Ter)	NF1	17	31343053	Neurofibromatosis, type 1
NM_000267.3(NF1):c.2848C>T (p.Gln950Ter)	NF1	17	31229463	Neurofibromatosis, type 1
NM_000267.3(NF1):c.79C>T (p.Gln27Ter)	NF1	17	31156001	Neurofibromatosis, type 1
NM_000267.3(NF1):c.6641+1G>A	NF1	17	31337881	Neurofibromatosis, type 1
NM_000267.3(NF1):c.4495C>T (p.Gln1499Ter)	NF1	17	31260496	Neurofibromatosis, type 1
NM_000267.3(NF1):c.4168C>T (p.Leu1390Phe)	NF1	17	31258401	Neurofibromatosis, type 1;Neurofibromatosis-Noonan syndrome;not provided
NM_000267.3(NF1):c.5839C>T (p.Arg1947Ter)	NF1	17	31334927	Neurofibromatosis, type 1;not provided
NM_000267.3(NF1):c.4084C>T (p.Arg1362Ter)	NF1	17	31249093	Neurofibromatosis, type 1;not provided
NM_000267.3(NF1):c.574C>T (p.Arg192Ter)	NF1	17	31169985	Neurofibromatosis, type 1;not provided
NM_001042492.2(NF1):c.3827G>A (p.Arg1276Gln)	NF1	17	31235729	Neurofibromatosis, type 1;not provided
NM_000267.3(NF1):c.5425C>T (p.Arg1809Cys)	NF1	17	31327718	Neurofibromatosis, type 1;not provided
NM_000267.3(NF1):c.5242C>T (p.Arg1748Ter)	NF1	17	31327535	Neurofibromatosis, type 1;not provided
NM_000267.3(NF1):c.1381C>T (p.Arg461Ter)	NF1	17	31206360	Neurofibromatosis, type 1;not provided
NM_000267.3(NF1):c.3826C>T (p.Arg1276Ter)	NF1	17	31235728	Neurofibromatosis, type 1;not provided
NM_000267.3(NF1):c.1726C>T (p.Gln576Ter)	NF1	17	31223448	Neurofibromatosis, type 1;not provided
NM_000267.3(NF1):c.1246C>T (p.Arg416Ter)	NF1	17	31201471	Neurofibromatosis, type 1;not provided
NM_000268.3(NF2):c.784C>T (p.Arg262Ter)	NF2	22	29661313	Neurofibromatosis, type 2
NM_000268.3(NF2):c.958C>T (p.Gln320Ter)	NF2	22	29668405	Neurofibromatosis, type 2
NM_000268.3(NF2):c.1021C>T (p.Arg341Ter)	NF2	22	29671847	Neurofibromatosis, type 2
NM_000268.3(NF2):c.1219C>T (p.Gln407Ter)	NF2	22	29673365	Neurofibromatosis, type 2
NM_000268.3(NF2):c.1396C>T (p.Arg466Ter)	NF2	22	29674891	Neurofibromatosis, type 2
NM_130439.3(MXI1):c.362C>T (p.Ala121Val)	MXI1	10	110228276	Neurofibrosarcoma
m.5532G>A	MT-TW	MT	5532	Neurogastrointestinal syndrome, mitochondrial
NM_000490.4(AVP):c.262G>A (p.Gly88Ser)	AVP	20	3083037	Neurohypophyseal diabetes insipidus
NM_000490.4(AVP):c.55G>A (p.Ala19Thr)	AVP	20	3084620	Neurohypophyseal diabetes insipidus
NM_000490.4(AVP):c.56C>T (p.Ala19Val)	AVP	20	3084619	Neurohypophyseal diabetes insipidus
NM_000490.4(AVP):c.260C>T (p.Ser87Phe)	AVP	20	3083039	Neurohypophyseal diabetes insipidus
NM_000490.4(AVP):c.275G>A (p.Cys92Tyr)	AVP	20	3083024	Neurohypophyseal diabetes insipidus
NM_021625.4(TRPV4):c.1781G>A (p.Arg594His)	TRPV4	12	109792695	Neuromuscular Diseases;Parastremmatic dwarfism;Skeletal dysplasia;Spondylometaphyseal dysplasia, Kozlowski type
NM_021625.4(TRPV4):c.832G>A (p.Glu278Lys)	TRPV4	12	109800639	Neuromuscular Diseases;Skeletal dysplasia;Spondylometaphyseal dysplasia, Kozlowski type
NM_005340.6(HINT1):c.184C>T (p.Gln62Ter)	HINT1	5	131162604	Neuromyotonia and axonal neuropathy, autosomal recessive
NM_005340.6(HINT1):c.278G>A (p.Gly93Asp)	HINT1	5	131159550	Neuromyotonia and axonal neuropathy, autosomal recessive
NM_006415.3(SPTLC1):c.398G>A (p.Cys133Tyr)	SPTLC1	9	92080045	Neuropathy hereditary sensory and autonomic type 1
NM_006415.3(SPTLC1):c.1055C>T (p.Ala352Val)	SPTLC1	9	92047198	Neuropathy hereditary sensory and autonomic type 1
NM_000530.7(MPZ):c.643C>T (p.Gln215Ter)	MPZ	1	161306110	Neuropathy, congenital hypomyelinating, autosomal dominant
NM_006070.5(TFG):c.854C>T (p.Pro285Leu)	TFG	3	100748182	Neuropathy, hereditary motor and sensory, Okinawa type;not provided
NM_138773.3(SLC25A46):c.746G>A (p.Gly249Asp)	SLC25A46	5	110761271	NEUROPATHY, HEREDITARY MOTOR AND SENSORY, TYPE VIB
NM_138773.3(SLC25A46):c.998C>T (p.Pro333Leu)	SLC25A46	5	110761523	NEUROPATHY, HEREDITARY MOTOR AND SENSORY, TYPE VIB
NM_138773.3(SLC25A46):c.1018C>T (p.Arg340Cys)	SLC25A46	5	110761543	NEUROPATHY, HEREDITARY MOTOR AND SENSORY, TYPE VIB
NM_138773.3(SLC25A46):c.425C>T (p.Thr142Ile)	SLC25A46	5	110746309	NEUROPATHY, HEREDITARY MOTOR AND SENSORY, TYPE VIB
NM_006415.3(SPTLC1):c.992C>T (p.Ser331Phe)	SPTLC1	9	92047261	Neuropathy, hereditary sensory and autonomic, type I, severe
NM_004863.3(SPTLC2):c.1075G>A (p.Val359Met)	SPTLC2	14	77555401	NEUROPATHY, HEREDITARY SENSORY, TYPE IC
NM_001003841.2(SLC6A19):c.718C>T (p.Arg240Ter)	SLC6A19	5	1213517	Neutral 1 amino acid transport defect

NM_001003841.2(SLC6A19):c.517G>A (p.Asp173Asn)	SLC6A19	5	1212338	Neutral 1 amino acid transport defect;not provided
NM_020376.3(PNPLA2):c.584C>T (p.Pro195Leu)	PNPLA2	11	822494	Neutral lipid storage disease with myopathy
NM_020376.3(PNPLA2):c.865C>T (p.Gln289Ter)	PNPLA2	11	823801	Neutral lipid storage disease with myopathy
NM_000760.3(CSF3R):c.922C>T (p.Arg308Cys)	CSF3R	1	36472313	Neutropenia, severe congenital, 7, autosomal recessive;Severe congenital neutropenia
NM_002872.4(RAC2):c.169G>A (p.Asp57Asn)	RAC2	22	37232857	Neutrophil immunodeficiency syndrome
NM_003070.4(SMARCA2):c.3395G>A (p.Gly1132Asp)	SMARCA2	9	2110356	Nicolaides-Baraitser syndrome
NM_017519.2(ARID1B):c.1735C>T (p.Gln579Ter)	ARID1B	6	156829419	Nicolaides-Baraitser syndrome
NM_003070.4(SMARCA2):c.3637C>T (p.Arg1213Trp)	SMARCA2	9	2116002	Nicolaides-Baraitser syndrome;not provided
NM_003070.4(SMARCA2):c.3476G>A (p.Arg1159Gln)	SMARCA2	9	2113841	Nicolaides-Baraitser syndrome;not provided
NM_003070.4(SMARCA2):c.3485G>A (p.Arg1162His)	SMARCA2	9	2115850	Nicolaides-Baraitser syndrome;not provided
NM_003070.4(SMARCA2):c.2648C>T (p.Pro883Leu)	SMARCA2	9	2086950	Nicolaides-Baraitser syndrome;not provided
NM_003070.4(SMARCA2):c.3602C>T (p.Ala1201Val)	SMARCA2	9	2115967	Nicolaides-Baraitser syndrome;not provided
NM_003070.4(SMARCA2):c.2815C>T (p.His939Tyr)	SMARCA2	9	2088545	Nicolaides-Baraitser syndrome;not provided
NM_000271.4(NPC1):c.2873G>A (p.Arg958Gln)	NPC1	18	23539393	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.3591+1G>A	NPC1	18	23534445	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.530G>A (p.Cys177Tyr)	NPC1	18	23561461	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.3160G>A (p.Ala1054Thr)	NPC1	18	23536758	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.1553G>A (p.Arg518Gln)	NPC1	18	23554758	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.2128C>T (p.Gln710Ter)	NPC1	18	23544346	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.2366G>A (p.Arg789His)	NPC1	18	23541313	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.3175C>T (p.Arg1059Ter)	NPC1	18	23536743	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.1142G>A (p.Trp381Ter)	NPC1	18	23556427	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.2872C>T (p.Arg958Ter)	NPC1	18	23539394	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.3229C>T (p.Arg1077Ter)	NPC1	18	23536689	Niemann-Pick disease type C1
NM_000271.4(NPC1):c.2848G>A (p.Val950Met)	NPC1	18	23539418	Niemann-Pick disease type C1;not provided
NM_000271.4(NPC1):c.3104C>T (p.Ala1035Val)	NPC1	18	23536814	Niemann-Pick disease type C1;not provided
NM_000271.4(NPC1):c.2932C>T (p.Arg978Cys)	NPC1	18	23538651	Niemann-Pick disease type C1;not provided
NM_000271.4(NPC1):c.2974G>A (p.Gly992Arg)	NPC1	18	23538609	Niemann-Pick disease type C1;not provided
NM_000271.4(NPC1):c.2861C>T (p.Ser954Leu)	NPC1	18	23539405	Niemann-Pick disease type C1;not provided
NM_000271.4(NPC1):c.3557G>A (p.Arg1186His)	NPC1	18	23534480	Niemann-Pick disease type C1;not provided
NM_000271.4(NPC1):c.1211G>A (p.Arg404Gln)	NPC1	18	23556358	Niemann-Pick disease type C1;not provided
NM_006432.3(NPC2):c.190+5G>A	NPC2	14	74486324	Niemann-Pick disease type C2
NM_006432.3(NPC2):c.115G>A (p.Val39Met)	NPC2	14	74486404	Niemann-Pick disease type C2
NM_006432.3(NPC2):c.436C>T (p.Gln146Ter)	NPC2	14	74480707	Niemann-Pick disease type C2
NM_006432.3(NPC2):c.358C>T (p.Pro120Ser)	NPC2	14	74484420	Niemann-Pick disease type C2
NM_006432.3(NPC2):c.133C>T (p.Gln45Ter)	NPC2	14	74486386	Niemann-Pick disease type C2
NM_000543.4(SMPD1):c.1735G>A (p.Gly579Ser)	SMPD1	11	6394446	Niemann-Pick disease, type A
NM_000543.4(SMPD1):c.1152G>A (p.Met384Ile)	SMPD1	11	6393276	Niemann-Pick disease, type A
NM_001007593.2(SMPD1):c.949G>A (p.Val317Met)	SMPD1	11	6392017	Niemann-Pick disease, type A
NM_000543.4(SMPD1):c.1327C>T (p.Arg443Ter)	SMPD1	11	6393680	Niemann-Pick disease, type A;Niemann-Pick disease, type B;not provided
NM_000543.4(SMPD1):c.1267C>T (p.His423Tyr)	SMPD1	11	6393620	Niemann-Pick disease, type A;Niemann-Pick disease, type B;Sphingomyelin/cholesterol lipidosis
NM_000543.4(SMPD1):c.1426C>T (p.Arg476Trp)	SMPD1	11	6393981	Niemann-Pick disease, type A;Niemann-Pick disease, type B;Sphingomyelin/cholesterol lipidosis;not provided
NM_000543.4(SMPD1):c.730G>A (p.Gly244Arg)	SMPD1	11	6391795	Niemann-Pick disease, type B
NM_000543.4(SMPD1):c.1624C>T (p.Arg542Ter)	SMPD1	11	6394335	Niemann-Pick disease, type B;not provided

NM_000271.4(NPC1):c.2665G>A (p.Val889Met)	NPC1	18	23539941	Niemann-Pick disease, type C1, adult form
NM_002075.3(GNB3):c.1017G>A (p.Trp339Ter)	GNB3	12	6846892	Night blindness, congenital stationary, type 1h
NM_002075.3(GNB3):c.200C>T (p.Ser67Phe)	GNB3	12	6843073	Night blindness, congenital stationary, type 1h
NM_005732.3(RAD50):c.3277C>T (p.Arg1093Ter)	RAD50	5	132618182	Nijmegen breakage syndrome-like disorder
NM_001128227.2(GNE):c.1820G>A (p.Gly607Glu)	GNE	9	36219927	Nonaka myopathy
NM_001190383.1(GNE):c.1669G>A (p.Ala557Thr)	GNE	9	36218225	Nonaka myopathy
NM_001128227.2(GNE):c.766G>A (p.Asp256Asn)	GNE	9	36236928	Nonaka myopathy
NM_005476.5(GNE):c.1571C>T (p.Ala524Val)	GNE	9	36222839	Nonaka myopathy
NM_003900.4(SQSTM1):c.970_1165del	SQSTM1	5	179833783	Nonaka myopathy;Paget disease of bone, familial
NM_000481.3(AMT):c.806G>A (p.Gly269Asp)	AMT	3	49419042	Non-ketotic hyperglycinemia
NM_000481.3(AMT):c.139G>A (p.Gly47Arg)	AMT	3	49422223	Non-ketotic hyperglycinemia
NM_000481.3(AMT):c.959G>A (p.Arg320His)	AMT	3	49417892	Non-ketotic hyperglycinemia
NM_000481.3(AMT):c.574C>T (p.Gln192Ter)	AMT	3	49419382	Non-ketotic hyperglycinemia
NM_000481.3(AMT):c.878-1G>A	AMT	3	49417974	Non-ketotic hyperglycinemia
NM_000170.2(GLDC):c.2405C>T (p.Ala802Val)	GLDC	9	6553420	Non-ketotic hyperglycinemia
NM_000170.2(GLDC):c.2216G>A (p.Arg739His)	GLDC	9	6554768	Non-ketotic hyperglycinemia
NM_000170.2(GLDC):c.2284G>A (p.Gly762Arg)	GLDC	9	6554700	Non-ketotic hyperglycinemia
NM_001164712.1(AMT):c.870G>A (p.Trp290Ter)	AMT	3	49418978	Non-ketotic hyperglycinemia
NM_000170.2(GLDC):c.1166C>T (p.Ala389Val)	GLDC	9	6595109	Non-ketotic hyperglycinemia;not provided
NM_000170.2(GLDC):c.1009C>T (p.Arg337Ter)	GLDC	9	6604637	Non-ketotic hyperglycinemia;not provided
NM_198173.2(GRHL3):c.738C>T (p.Gly246=)	GRHL3	1	24337687	nonsyndromic cleft palate
NM_000307.4(POU3F4):c.499C>T (p.Arg167Ter)	POU3F4	X	83508823	Nonsyndromic hearing loss and deafness
NM_004004.5(GJB2):c.169C>T (p.Gln57Ter)	GJB2	13	20189413	Nonsyndromic hearing loss and deafness
NM_016239.3(MYO15A):c.8767C>T (p.Arg2923Ter)	MYO15A	17	18157209	Nonsyndromic hearing loss and deafness
NM_024022.2(TMPRSS3):c.323-6G>A	TMPRSS3	21	42388532	Nonsyndromic hearing loss and deafness
NM_024022.2(TMPRSS3):c.916G>A (p.Ala306Thr)	TMPRSS3	21	42382101	Nonsyndromic hearing loss and deafness
NM_144612.6(LOXHD1):c.2497C>T (p.Arg833Ter)	LOXHD1	18	46563166	Nonsyndromic hearing loss and deafness
NM_194248.2(OTOF):c.2153G>A (p.Trp718Ter)	OTOF	2	26479325	Nonsyndromic hearing loss and deafness
NM_194248.2(OTOF):c.2818C>T (p.Gln940Ter)	OTOF	2	26476176	Nonsyndromic hearing loss and deafness
NM_194248.2(OTOF):c.4799+1G>A	OTOF	2	26465671	Nonsyndromic hearing loss and deafness
NM_004999.3(MYO6):c.826C>T (p.Arg276Ter)	MYO6	6	75844906	Nonsyndromic hearing loss and deafness
NM_144672.3(OTOA):c.1880+1G>A	OTOA	16	21722979	Nonsyndromic hearing loss and deafness
NM_153700.2(STRC):c.5188C>T (p.Arg1730Ter)	STRC	15	43600011	Nonsyndromic hearing loss and deafness
NM_153700.2(STRC):c.3670C>T (p.Arg1224Ter)	STRC	15	43608091	Nonsyndromic hearing loss and deafness
NM_153700.2(STRC):c.4402C>T (p.Arg1468Ter)	STRC	15	43603385	Nonsyndromic hearing loss and deafness
NM_024022.2(TMPRSS3):c.1192C>T (p.Gln398Ter)	TMPRSS3	21	42376543	Nonsyndromic hearing loss and deafness
NM_001039141.2(TRIOBP):c.6598C>T (p.Arg2200Ter)	TRIOBP	22	37769050	Nonsyndromic hearing loss and deafness
NM_016239.3(MYO15A):c.7893+1G>A	MYO15A	17	18151952	Nonsyndromic hearing loss and deafness
NM_016239.3(MYO15A):c.5531+1G>A	MYO15A	17	18141144	Nonsyndromic hearing loss and deafness
NM_016239.3(MYO15A):c.6046+1G>A	MYO15A	17	18143797	Nonsyndromic hearing loss and deafness
NM_144612.6(LOXHD1):c.3169C>T (p.Arg1057Ter)	LOXHD1	18	46559495	Nonsyndromic hearing loss and deafness
NM_001038603.2(MARVELD2):c.1331+1G>A	MARVELD2	5	69432676	Nonsyndromic hearing loss and deafness
NM_138691.2(TMC1):c.1676G>A (p.Trp559Ter)	TMC1	9	72805491	Nonsyndromic hearing loss and deafness
NM_138691.2(TMC1):c.1677G>A (p.Trp559Ter)	TMC1	9	72805492	Nonsyndromic hearing loss and deafness
NM_005422.2(TECTA):c.5977C>T (p.Arg1993Ter)	TECTA	11	121168903	Nonsyndromic hearing loss and deafness

NM_173591.3(OTOGL):c.4987C>T (p.Arg1663Ter)	OTOGL	12	80339228	Nonsyndromic hearing loss and deafness
NM_153700.2(STRC):c.3493C>T (p.Gln1165Ter)	STRC	15	43610317	Nonsyndromic hearing loss and deafness
NM_153700.2(STRC):c.3217C>T (p.Arg1073Ter)	STRC	15	43611237	Nonsyndromic hearing loss and deafness
NM_016239.3(MYO15A):c.5896C>T (p.Arg1966Ter)	MYO15A	17	18142826	Nonsyndromic hearing loss and deafness
NM_133261.2(GIPC3):c.411+1G>A	GIPC3	19	3586681	Nonsyndromic hearing loss and deafness
NM_001039213.3(CEACAM16):c.703C>T (p.Arg235Cys)	CEACAM16	19	44705631	Nonsyndromic hearing loss and deafness
NM_005633.3(SOS1):c.1300G>A (p.Gly434Arg)	SOS1	2	39023128	Noonan syndrome
NM_002834.4(PTPN11):c.1232C>T (p.Thr411Met)	PTPN11	12	112486482	Noonan syndrome 1
NM_002524.4(NRAS):c.179G>A (p.Gly60Glu)	NRAS	1	114713911	Noonan syndrome 1;Noonan syndrome 6;not provided
NM_006767.3(LZTR1):c.850C>T (p.Arg284Cys)	LZTR1	22	20991686	Noonan syndrome 10
NM_006767.3(LZTR1):c.740G>A (p.Ser247Asn)	LZTR1	22	20990474	Noonan syndrome 10
NM_006767.3(LZTR1):c.742G>A (p.Gly248Arg)	LZTR1	22	20990476	Noonan syndrome 10;not provided
NM_004985.4(KRAS):c.178G>A (p.Gly60Ser)	KRAS	12	25227346	Noonan syndrome 3;not provided
NM_002880.3(RAF1):c.1456G>A (p.Asp486Asn)	RAF1	3	12585761	Noonan syndrome with multiple lentigines
NM_002524.4(NRAS):c.149C>T (p.Thr50Ile)	NRAS	1	114713941	Noonan syndrome;Noonan syndrome 1;Noonan syndrome 6;Rasopathy
NM_004333.4(BRAF):c.722C>T (p.Thr241Met)	BRAF	7	140801550	Noonan syndrome;Noonan syndrome 1;Noonan syndrome 7;not provided
NM_002834.4(PTPN11):c.5C>T (p.Thr2Ile)	PTPN11	12	112419116	Noonan syndrome;Noonan syndrome 1;not provided
NM_002834.4(PTPN11):c.218C>T (p.Thr73Ile)	PTPN11	12	112450398	Noonan syndrome;Noonan syndrome 1;Rasopathy;not provided
NM_004985.4(KRAS):c.173C>T (p.Thr58Ile)	KRAS	12	25227351	Noonan syndrome;Noonan syndrome 3;not provided
NM_005633.3(SOS1):c.1655G>A (p.Arg552Lys)	SOS1	2	39022773	Noonan syndrome;Noonan syndrome 4;not provided
NM_002880.3(RAF1):c.782C>T (p.Pro261Leu)	RAF1	3	12604188	Noonan syndrome;Noonan syndrome 5
NM_002880.3(RAF1):c.781C>T (p.Pro261Ser)	RAF1	3	12604189	Noonan syndrome;Noonan syndrome 5;Primary familial hypertrophic cardiomyopathy;Rasopathy;not provided
NM_006912.5(RIT1):c.270G>A (p.Met90Ile)	RIT1	1	155904470	Noonan syndrome;Noonan syndrome 8;not provided
NM_002880.3(RAF1):c.1472C>T (p.Thr491Ile)	RAF1	3	12585745	Noonan syndrome;Noonan syndrome with multiple lentigines;not provided
NM_002834.4(PTPN11):c.178G>A (p.Gly60Ser)	PTPN11	12	112450358	Noonan syndrome;not provided
NM_002834.4(PTPN11):c.328G>A (p.Glu110Lys)	PTPN11	12	112450508	Noonan syndrome;not provided
NM_002834.4(PTPN11):c.781C>T (p.Leu261Phe)	PTPN11	12	112472968	Noonan syndrome;not provided
NM_002834.4(PTPN11):c.1505C>T (p.Ser502Leu)	PTPN11	12	112489081	Noonan syndrome;not provided
NM_005633.3(SOS1):c.322G>A (p.Glu108Lys)	SOS1	2	39058696	Noonan syndrome;not provided
NM_005633.3(SOS1):c.1322G>A (p.Cys441Tyr)	SOS1	2	39023106	Noonan syndrome;not provided
NM_006912.5(RIT1):c.229G>A (p.Ala77Thr)	RIT1	1	155904739	Noonan syndrome;not provided
NM_002880.3(RAF1):c.776C>T (p.Ser259Phe)	RAF1	3	12604194	Noonan syndrome;Rasopathy
NM_004985.4(KRAS):c.101C>T (p.Pro34Leu)	KRAS	12	25245284	Noonan syndrome;Rasopathy;not provided
NM_002834.4(PTPN11):c.1471C>T (p.Pro491Ser)	PTPN11	12	112489047	Noonan syndrome;Rasopathy;not provided
NM_002834.4(PTPN11):c.1472C>T (p.Pro491Leu)	PTPN11	12	112489048	Noonan syndrome;Rasopathy;not provided
NM_002834.4(PTPN11):c.1502G>A (p.Arg501Lys)	PTPN11	12	112489078	Noonan syndrome;Rasopathy;not provided
NM_002834.4(PTPN11):c.1507G>A (p.Gly503Arg)	PTPN11	12	112489083	Noonan syndrome;Rasopathy;not provided
NM_005633.3(SOS1):c.1297G>A (p.Glu433Lys)	SOS1	2	39023131	Noonan syndrome;Rasopathy;not provided
NM_005633.3(SOS1):c.2536G>A (p.Glu846Lys)	SOS1	2	39007168	Noonan syndrome;Rasopathy;not provided
NM_005188.3(CBL):c.1259G>A (p.Arg420Gln)	CBL	11	119278541	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia;not provided
NM_000229.1(LCAT):c.953G>A (p.Met317Ile)	LCAT	16	67940276	Norum disease
NM_000229.1(LCAT):c.475C>T (p.Arg159Trp)	LCAT	16	67942719	Norum disease
NM_000229.1(LCAT):c.1034C>T (p.Thr345Met)	LCAT	16	67940193	Norum disease
NM_004693.2(KRT75):c.481G>A (p.Ala161Thr)	KRT75	12	52433824	not provided

NM_023110.2(FGFR1):c.1825C>T (p.Arg609Ter)	FGFR1	8	38415899	not provided	
NM_020630.4(RET):c.1832G>A (p.Cys611Tyr)	RET	10	43113628	not provided	
NM_000942.4(PPIB):c.343+1G>A	PPIB	15	64160103	not provided	
NM_173630.3(RTTN):c.80G>A (p.Cys27Tyr)	RTTN	18	70205267	not provided	
NM_004333.4(BRAF):c.1513C>T (p.Leu505Phe)	BRAF	7	140777995	not provided	
NM_005343.3(HRAS):c.179G>A (p.Gly60Asp)	HRAS	11	533877	not provided	
NM_002834.4(PTPN11):c.1508G>A (p.Gly503Glu)	PTPN11	12	112489084	not provided	
NM_022173.2(TIA1):c.1150G>A (p.Glu384Lys)	TIA1	2	70212730	not provided	
NM_023110.2(FGFR1):c.1042G>A (p.Gly348Arg)	FGFR1	8	38421836	not provided	
NM_002016.1(FLG):c.7339C>T (p.Arg2447Ter)	FLG	1	152307547	not provided	
NM_000130.4(F5):c.5392G>A (p.Glu1798Lys)	F5	1	169529635	not provided	
NM_000130.4(F5):c.5668G>A (p.Glu1890Lys)	F5	1	169525949	not provided	
NM_000424.3(KRT5):c.1411C>T (p.Arg471Cys)	KRT5	12	52516665	not provided	
NM_000424.3(KRT5):c.502G>A (p.Glu168Lys)	KRT5	12	52519795	not provided	
NM_000526.4(KRT14):c.1228C>T (p.Gln410Ter)	KRT14	17	41583281	not provided	
NM_000526.4(KRT14):c.1231G>A (p.Glu411Lys)	KRT14	17	41583278	not provided	
NM_000526.4(KRT14):c.397G>A (p.Val133Met)	KRT14	17	41586438	not provided	
NM_000526.4(KRT14):c.915G>A (p.Trp305Ter)	KRT14	17	41583772	not provided	
NM_001927.3(DES):c.1237G>A (p.Glu413Lys)	DES	2	219421553	not provided	
NM_005554.3(KRT6A):c.487G>A (p.Glu163Lys)	KRT6A	12	52492702	not provided	
NM_005554.3(KRT6A):c.508C>T (p.Leu170Phe)	KRT6A	12	52492681	not provided	
NM_005555.3(KRT6B):c.1381G>A (p.Glu461Lys)	KRT6B	12	52447821	not provided	
NM_006121.3(KRT1):c.559C>T (p.Leu187Phe)	KRT1	12	52679790	not provided	
NM_006121.3(KRT1):c.698C>T (p.Ser233Leu)	KRT1	12	52678650	not provided	
NM_006158.4(NEFL):c.1186G>A (p.Glu396Lys)	NEFL	8	24953779	not provided	
NM_170707.3(LMNA):c.1045C>T (p.Arg349Trp)	LMNA	1	156136009	not provided	
NM_170707.3(LMNA):c.1380+1G>A	LMNA	1	156136437	not provided	
NM_170707.3(LMNA):c.412G>A (p.Glu138Lys)	LMNA	1	156130672	not provided	
NM_170707.3(LMNA):c.569G>A (p.Arg190Gln)	LMNA	1	156134458	not provided	
NM_170707.3(LMNA):c.736C>T (p.Gln246Ter)	LMNA	1	156134901	not provided	
NM_017890.4(VPS13B):c.1504C>T (p.Arg502Ter)	VPS13B	8	99135674	not provided	
NM_017890.4(VPS13B):c.1563G>A (p.Lys521=)	VPS13B	8	99135733	not provided	
NM_017890.4(VPS13B):c.5920C>T (p.Arg1974Ter)	VPS13B	8	99642435	not provided	
NM_017890.4(VPS13B):c.8318C>T (p.Ser2773Leu)	VPS13B	8	99817685	not provided	
NM_000397.3(CYBB):c.1223G>A (p.Gly408Glu)	CYBB	X	37805077	not provided	
NM_000448.2(RAG1):c.2924G>A (p.Arg975Gln)	RAG1	11	36576228	not provided	
NM_000693.3(ALDH1A3):c.211G>A (p.Val71Met)	ALDH1A3	15	100887578	not provided	
NM_000048.3(ASL):c.602+1G>A	ASL	7	66086822	not provided	
NM_000117.2(EMD):c.355C>T (p.Gln119Ter)	EMD	X	154380323	not provided	
NM_000143.3(FH):c.760C>T (p.Gln254Ter)	FH	1	241506147	not provided	
NM_000194.2(HPRT1):c.486-1G>A	HPRT1	X	134498389	not provided	
NM_000263.3(NAGLU):c.503G>A (p.Trp168Ter)	NAGLU	17	42537517	not provided	
NM_000266.3(NDP):c.109C>T (p.Arg37Ter)	NDP	X	43958537	not provided	
NM_000284.3(PDHA1):c.483C>T (p.Tyr161=)	PDHA1	X	19353146	not provided	
NM_000297.3(PKD2):c.709+1G>A	PKD2	4	88019572	not provided	

NM_000466.2(PEX1):c.2230C>T (p.Gln744Ter)	PEX1	7	92502076	not provided	
NM_000489.4(ATRX):c.189+1G>A	ATRX	X	77698573	not provided	
NM_000528.3(MAN2B1):c.1929G>A (p.Trp643Ter)	MAN2B1	19	12652270	not provided	
NM_000528.3(MAN2B1):c.2398G>A (p.Gly800Arg)	MAN2B1	19	12649174	not provided	
NM_001143979.1(NDE1):c.704-1G>A	NDE1	16	15694164	not provided	
NM_003361.3(UMOD):c.944G>A (p.Cys315Tyr)	UMOD	16	20348252	not provided	
NM_003919.2(SGCE):c.709C>T (p.Arg237Ter)	SGCE	7	94603406	not provided	
NM_004380.2(CREBBP):c.3337C>T (p.Gln1113Ter)	CREBBP	16	3758886	not provided	
NM_004380.2(CREBBP):c.3982+1G>A	CREBBP	16	3744893	not provided	
NM_004737.5(LARGE1):c.1102C>T (p.Gln368Ter)	LARGE1	22	33381948	not provided	
NM_001042537.1(SLC9A6):c.1072C>T (p.Gln358Ter)	SLC9A6	X	136012979	not provided	
NM_032985.4(SEC23B):c.1571C>T (p.Ala524Val)	SEC23B	20	18543078	not provided	
NM_032985.4(SEC23B):c.689+1G>A	SEC23B	20	18525021	not provided	
NM_013382.5(POMT2):c.1417C>T (p.Arg473Ter)	POMT2	14	77285548	not provided	
NM_014141.5(CNTNAP2):c.2153G>A (p.Trp718Ter)	CNTNAP2	7	147903619	not provided	
NM_019109.4(ALG1):c.1187+1G>A	ALG1	16	5082674	not provided	
NM_020366.3(RPGRIP1):c.2775G>A (p.Trp925Ter)	RPGRIP1	14	21327687	not provided	
NM_024079.4(ALG8):c.121C>T (p.Arg41Ter)	ALG8	11	78127411	not provided	
NM_130799.2(MEN1):c.912+1G>A	MEN1	11	64807010	not provided	
NM_144997.5(FLCN):c.1060C>T (p.Gln354Ter)	FLCN	17	17219021	not provided	
NM_144997.5(FLCN):c.1597C>T (p.Gln533Ter)	FLCN	17	17213798	not provided	
NM_152419.2(HGSNAT):c.1250+1G>A	HGSNAT	8	43191596	not provided	
NM_152419.2(HGSNAT):c.1464+1G>A	HGSNAT	8	43193844	not provided	
NM_198270.3(NHS):c.953G>A (p.Trp318Ter)	NHS	X	17721541	not provided	
NM_000531.5(OTC):c.1005G>A (p.Met335Ile)	OTC	X	38411999	not provided	
NM_000531.5(OTC):c.1006-1G>A	OTC	X	38421022	not provided	
NM_000531.5(OTC):c.1042C>T (p.Gln348Ter)	OTC	X	38421059	not provided	
NM_000531.5(OTC):c.106C>T (p.Gln36Ter)	OTC	X	38367319	not provided	
NM_000531.5(OTC):c.127C>T (p.Leu43Phe)	OTC	X	38367340	not provided	
NM_000531.5(OTC):c.131C>T (p.Thr44Ile)	OTC	X	38367344	not provided	
NM_000531.5(OTC):c.148G>A (p.Gly50Arg)	OTC	X	38367361	not provided	
NM_000531.5(OTC):c.154G>A (p.Glu52Lys)	OTC	X	38367367	not provided	
NM_000531.5(OTC):c.174G>A (p.Trp58Ter)	OTC	X	38367387	not provided	
NM_000531.5(OTC):c.179C>T (p.Ser60Leu)	OTC	X	38367392	not provided	
NM_000531.5(OTC):c.205C>T (p.Gln69Ter)	OTC	X	38367418	not provided	
NM_000531.5(OTC):c.216+1G>A	OTC	X	38367430	not provided	
NM_000531.5(OTC):c.217-1G>A	OTC	X	38369795	not provided	
NM_000531.5(OTC):c.232C>T (p.Gln78Ter)	OTC	X	38369811	not provided	
NM_000531.5(OTC):c.248G>A (p.Gly83Asp)	OTC	X	38369827	not provided	
NM_000531.5(OTC):c.269G>A (p.Ser90Asn)	OTC	X	38369848	not provided	
NM_000531.5(OTC):c.274C>T (p.Arg92Ter)	OTC	X	38369853	not provided	
NM_000531.5(OTC):c.275G>A (p.Arg92Gln)	OTC	X	38369854	not provided	
NM_000531.5(OTC):c.292G>A (p.Glu98Lys)	OTC	X	38369871	not provided	
NM_000531.5(OTC):c.298+1G>A	OTC	X	38369878	not provided	
NM_000531.5(OTC):c.299G>A (p.Gly100Asp)	OTC	X	38381342	not provided	

NM_000531.5(OTC):c.316G>A (p.Gly106Arg)	OTC	X	38381359	not provided
NM_000531.5(OTC):c.317G>A (p.Gly106Glu)	OTC	X	38381360	not provided
NM_000531.5(OTC):c.386+1G>A	OTC	X	38381430	not provided
NM_000531.5(OTC):c.395C>T (p.Ser132Phe)	OTC	X	38401283	not provided
NM_000531.5(OTC):c.3G>A (p.Met1Ile)	OTC	X	38352699	not provided
NM_000531.5(OTC):c.409G>A (p.Ala137Thr)	OTC	X	38401297	not provided
NM_000531.5(OTC):c.455C>T (p.Ala152Val)	OTC	X	38401343	not provided
NM_000531.5(OTC):c.485G>A (p.Gly162Glu)	OTC	X	38401373	not provided
NM_000531.5(OTC):c.506C>T (p.Pro169Leu)	OTC	X	38401394	not provided
NM_000531.5(OTC):c.533C>T (p.Thr178Met)	OTC	X	38401421	not provided
NM_000531.5(OTC):c.571C>T (p.Leu191Phe)	OTC	X	38403648	not provided
NM_000531.5(OTC):c.578G>A (p.Trp193Ter)	OTC	X	38403655	not provided
NM_000531.5(OTC):c.583G>A (p.Gly195Arg)	OTC	X	38403660	not provided
NM_000531.5(OTC):c.586G>A (p.Asp196Asn)	OTC	X	38403663	not provided
NM_000531.5(OTC):c.589G>A (p.Gly197Arg)	OTC	X	38403666	not provided
NM_000531.5(OTC):c.590G>A (p.Gly197Glu)	OTC	X	38403667	not provided
NM_000531.5(OTC):c.604C>T (p.His202Tyr)	OTC	X	38403681	not provided
NM_000531.5(OTC):c.620G>A (p.Ser207Asn)	OTC	X	38403697	not provided
NM_000531.5(OTC):c.622G>A (p.Ala208Thr)	OTC	X	38403699	not provided
NM_000531.5(OTC):c.626C>T (p.Ala209Val)	OTC	X	38403703	not provided
NM_000531.5(OTC):c.640C>T (p.His214Tyr)	OTC	X	38403717	not provided
NM_000531.5(OTC):c.643C>T (p.Leu215Phe)	OTC	X	38403720	not provided
NM_000531.5(OTC):c.659C>T (p.Pro220Leu)	OTC	X	38403736	not provided
NM_000531.5(OTC):c.663+1G>A	OTC	X	38403741	not provided
NM_000531.5(OTC):c.663G>A (p.Lys221=)	OTC	X	38403740	not provided
NM_000531.5(OTC):c.664-1G>A	OTC	X	38408741	not provided
NM_000531.5(OTC):c.67C>T (p.Arg23Ter)	OTC	X	38352763	not provided
NM_000531.5(OTC):c.698C>T (p.Ala233Val)	OTC	X	38408776	not provided
NM_000531.5(OTC):c.717+1G>A	OTC	X	38408796	not provided
NM_000531.5(OTC):c.717G>A (p.Glu239=)	OTC	X	38408795	not provided
NM_000531.5(OTC):c.725C>T (p.Thr242Ile)	OTC	X	38408883	not provided
NM_000531.5(OTC):c.757G>A (p.Ala253Thr)	OTC	X	38408915	not provided
NM_000531.5(OTC):c.77+1G>A	OTC	X	38352774	not provided
NM_000531.5(OTC):c.77+5G>A	OTC	X	38352778	not provided
NM_000531.5(OTC):c.785C>T (p.Thr262Ile)	OTC	X	38408943	not provided
NM_000531.5(OTC):c.787G>A (p.Asp263Asn)	OTC	X	38408945	not provided
NM_000531.5(OTC):c.791C>T (p.Thr264Ile)	OTC	X	38408949	not provided
NM_000531.5(OTC):c.795G>A (p.Trp265Ter)	OTC	X	38408953	not provided
NM_000531.5(OTC):c.806G>A (p.Gly269Glu)	OTC	X	38408964	not provided
NM_000531.5(OTC):c.808C>T (p.Gln270Ter)	OTC	X	38408966	not provided
NM_000531.5(OTC):c.830G>A (p.Arg277Gln)	OTC	X	38408988	not provided
NM_000531.5(OTC):c.835C>T (p.Gln279Ter)	OTC	X	38408993	not provided
NM_000531.5(OTC):c.867+1G>A	OTC	X	38409026	not provided
NM_000531.5(OTC):c.904C>T (p.His302Tyr)	OTC	X	38411898	not provided
NM_000531.5(OTC):c.908G>A (p.Cys303Tyr)	OTC	X	38411902	not provided

NM_000531.5(OTC):c.931G>A (p.Val311Met)	OTC	X	38411925	not provided
NM_000531.5(OTC):c.94C>T (p.Gln32Ter)	OTC	X	38367307	not provided
NM_000531.5(OTC):c.953C>T (p.Ser318Phe)	OTC	X	38411947	not provided
NM_000531.5(OTC):c.958C>T (p.Arg320Ter)	OTC	X	38411952	not provided
NM_000531.5(OTC):c.976G>A (p.Glu326Lys)	OTC	X	38411970	not provided
NM_000531.5(OTC):c.995G>A (p.Trp332Ter)	OTC	X	38411989	not provided
NM_000531.5(OTC):c.996G>A (p.Trp332Ter)	OTC	X	38411990	not provided
NM_000180.3(GUCY2D):c.2302C>T (p.Arg768Trp)	GUCY2D	17	8013918	not provided
NM_000322.4(PRPH2):c.715C>T (p.Gln239Ter)	PRPH2	6	42704478	not provided
NM_000330.3(RS1):c.599G>A (p.Arg200His)	RS1	X	18642080	not provided
NM_000330.3(RS1):c.626G>A (p.Arg209His)	RS1	X	18642053	not provided
NM_000330.3(RS1):c.637C>T (p.Arg213Trp)	RS1	X	18642042	not provided
NM_000350.2(ABCA4):c.1317G>A (p.Trp439Ter)	ABCA4	1	94078629	not provided
NM_000350.2(ABCA4):c.2948C>T (p.Thr983Ile)	ABCA4	1	94044715	not provided
NM_000350.2(ABCA4):c.5316G>A (p.Trp1772Ter)	ABCA4	1	94014687	not provided
NM_000350.2(ABCA4):c.5929G>A (p.Gly1977Ser)	ABCA4	1	94007710	not provided
NM_004183.3(BEST1):c.272C>T (p.Thr91Ile)	BEST1	11	61955742	not provided
NM_004183.3(BEST1):c.920C>T (p.Thr307Ile)	BEST1	11	61959550	not provided
NM_004183.3(BEST1):c.934G>A (p.Asp312Asn)	BEST1	11	61959564	not provided
NM_000552.4(VWF):c.4135C>T (p.Arg1379Cys)	VWF	12	6019283	not provided
NM_000277.1(PAH):c.722G>A (p.Arg241His)	PAH	12	102852935	not provided
NM_000038.5(APC):c.5314+5G>A	APC	5	112775742	not provided
NM_000051.3(ATM):c.1235G>A (p.Trp412Ter)	ATM	11	108249102	not provided
NM_000051.3(ATM):c.3931C>T (p.Gln1311Ter)	ATM	11	108284411	not provided
NM_000179.2(MSH6):c.1241G>A (p.Trp414Ter)	MSH6	2	47799224	not provided
NM_000179.2(MSH6):c.3142C>T (p.Gln1048Ter)	MSH6	2	47801125	not provided
NM_000251.2(MSH2):c.70C>T (p.Gln24Ter)	MSH2	2	47403261	not provided
NM_000465.3(BARD1):c.3G>A (p.Met1Ile)	BARD1	2	214809567	not provided
NM_000540.2(RYR1):c.14210G>A (p.Arg4737Gln)	RYR1	19	38577955	not provided
NM_000540.2(RYR1):c.7063C>T (p.Arg2355Trp)	RYR1	19	38499670	not provided
NM_000192.3(TBX5):c.338G>A (p.Arg113Lys)	TBX5	12	114399537	not provided
NM_005857.4(ZMPSTE24):c.1204-1G>A	ZMPSTE24	1	40292444	not provided
NM_004992.3(MECP2):c.46C>T (p.Gln16Ter)	MECP2	X	154032538	not provided
NM_024675.3(PALB2):c.1108C>T (p.Gln370Ter)	PALB2	16	23635438	not provided
NM_024675.3(PALB2):c.2074C>T (p.Gln692Ter)	PALB2	16	23630080	not provided
NM_024675.3(PALB2):c.48G>A (p.Lys16=)	PALB2	16	23641110	not provided
NM_003159.2(CDKL5):c.413C>T (p.Pro138Leu)	CDKL5	X	18581900	not provided
NM_003159.2(CDKL5):c.577G>A (p.Asp193Asn)	CDKL5	X	18587976	not provided
NM_003159.2(CDKL5):c.700C>T (p.Gln234Ter)	CDKL5	X	18588099	not provided
NM_139058.2(ARX):c.1141G>A (p.Ala381Thr)	ARX	X	25007418	not provided
NM_003159.2(CDKL5):c.73G>A (p.Gly25Arg)	CDKL5	X	18510828	not provided
NM_004992.3(MECP2):c.568C>T (p.Arg190Cys)	MECP2	X	154031260	not provided
NM_003159.2(CDKL5):c.1324C>T (p.Gln442Ter)	CDKL5	X	18604248	not provided
NM_003159.2(CDKL5):c.2671C>T (p.Gln891Ter)	CDKL5	X	18628545	not provided
NM_139058.2(ARX):c.1111C>T (p.Arg371Ter)	ARX	X	25010268	not provided

NM_000540.2(RYR1):c.4225C>T (p.Arg1409Ter)	RYR1	19	38475382	not provided	
NM_002206.2(ITGA7):c.2357+1G>A	ITGA7	12	55694442	not provided	
NM_032985.4(SEC23B):c.1648C>T (p.Arg550Ter)	SEC23B	20	18543155	not provided	
NM_001043353.1(TPM3):c.436C>T (p.Arg146Ter)	TPM3	1	154172927	not provided	
NM_000046.3(ARSB):c.944G>A (p.Arg315Gln)	ARSB	5	78885782	not provided	
NM_032985.4(SEC23B):c.1489C>T (p.Arg497Cys)	SEC23B	20	18542380	not provided	
NM_033018.3(CDK16):c.1258C>T (p.Arg420Ter)	CDK16	X	47227098	not provided	
NM_080629.2(COL11A1):c.3745-1G>A	COL11A1	1	102920365	not provided	
NM_001001430.2(TNNT2):c.861G>A (p.Trp287Ter)	TNNT2	1	201359216	not provided	
NM_001001430.2(TNNT2):c.422G>A (p.Arg141Gln)	TNNT2	1	201364335	not provided	
NM_001001430.2(TNNT2):c.239C>T (p.Pro80Leu)	TNNT2	1	201365635	not provided	
NM_005633.3(SOS1):c.1270G>A (p.Glu424Lys)	SOS1	2	39023158	not provided	
NM_003280.2(TNNC1):c.91G>A (p.Ala31Thr)	TNNC1	3	52452217	not provided	
NM_000256.3(MYBPC3):c.3775C>T (p.Gln1259Ter)	MYBPC3	11	47332111	not provided	
NM_000256.3(MYBPC3):c.3553C>T (p.Gln1185Ter)	MYBPC3	11	47332640	not provided	
NM_000256.3(MYBPC3):c.3034C>T (p.Gln1012Ter)	MYBPC3	11	47333713	not provided	
NM_000256.3(MYBPC3):c.3021G>A (p.Trp1007Ter)	MYBPC3	11	47333726	not provided	
NM_000256.3(MYBPC3):c.2995-1G>A	MYBPC3	11	47333753	not provided	
NM_000256.3(MYBPC3):c.2992C>T (p.Gln998Ter)	MYBPC3	11	47333924	not provided	
NM_000256.3(MYBPC3):c.2893C>T (p.Gln965Ter)	MYBPC3	11	47335054	not provided	
NM_000256.3(MYBPC3):c.2748G>A (p.Trp916Ter)	MYBPC3	11	47335199	not provided	
NM_000256.3(MYBPC3):c.2414G>A (p.Gly805Asp)	MYBPC3	11	47337579	not provided	
NM_000256.3(MYBPC3):c.1898-1G>A	MYBPC3	11	47341033	not provided	
NM_000256.3(MYBPC3):c.1731G>A (p.Trp577Ter)	MYBPC3	11	47342050	not provided	
NM_000256.3(MYBPC3):c.1624+5G>A	MYBPC3	11	47342573	not provided	
NM_000256.3(MYBPC3):c.1522C>T (p.Gln508Ter)	MYBPC3	11	47342680	not provided	
NM_000256.3(MYBPC3):c.1457G>A (p.Trp486Ter)	MYBPC3	11	47342830	not provided	
NM_000256.3(MYBPC3):c.1405C>T (p.Gln469Ter)	MYBPC3	11	47342882	not provided	
NM_000256.3(MYBPC3):c.1387C>T (p.Gln463Ter)	MYBPC3	11	47342900	not provided	
NM_000256.3(MYBPC3):c.1223+1G>A	MYBPC3	11	47343491	not provided	
NM_000256.3(MYBPC3):c.1201C>T (p.Gln401Ter)	MYBPC3	11	47343514	not provided	
NM_000256.3(MYBPC3):c.1188G>A (p.Trp396Ter)	MYBPC3	11	47343527	not provided	
NM_000256.3(MYBPC3):c.1120C>T (p.Gln374Ter)	MYBPC3	11	47343595	not provided	
NM_000256.3(MYBPC3):c.1015C>T (p.Gln339Ter)	MYBPC3	11	47346282	not provided	
NM_000256.3(MYBPC3):c.505+1G>A	MYBPC3	11	47350013	not provided	
NM_000257.3(MYH7):c.5410G>A (p.Ala1804Thr)	MYH7	14	23415144	not provided	
NM_000257.3(MYH7):c.4807G>A (p.Ala1603Thr)	MYH7	14	23416150	not provided	
NM_000257.3(MYH7):c.4144C>T (p.Arg1382Trp)	MYH7	14	23418235	not provided	
NM_000257.3(MYH7):c.2761G>A (p.Glu921Lys)	MYH7	14	23424068	not provided	
NM_000257.3(MYH7):c.2347C>T (p.Arg783Cys)	MYH7	14	23425358	not provided	
NM_000257.3(MYH7):c.1325G>A (p.Arg442His)	MYH7	14	23429037	not provided	
NM_000257.3(MYH7):c.610C>T (p.Arg204Cys)	MYH7	14	23431790	not provided	
NM_005159.4(ACTC1):c.968C>T (p.Ala323Val)	ACTC1	15	34791136	not provided	
NM_000371.3(TTR):c.130C>T (p.Pro44Ser)	TTR	18	31592956	not provided	
NM_000169.2(GLA):c.1085C>T (p.Pro362Leu)	GLA	X	101398014	not provided	

NM_000169.2(GLA):c.801G>A (p.Met267Ile)	GLA	X	101398785	not provided	
NM_000169.2(GLA):c.713G>A (p.Ser238Asn)	GLA	X	101398873	not provided	
NM_000169.2(GLA):c.548-1G>A	GLA	X	101400758	not provided	
NM_002294.2(LAMP2):c.864+1G>A	LAMP2	X	120446304	not provided	
NM_000249.3(MLH1):c.2065C>T (p.Gln689Ter)	MLH1	3	37048979	not provided	
NM_024675.3(PALB2):c.2834+1G>A	PALB2	16	23624008	not provided	
NM_007194.3(CHEK2):c.1356G>A (p.Trp452Ter)	CHEK2	22	28695146	not provided	
NM_001453.2(FOXC1):c.-244C>T	FOXC1	6	1610202	not provided	
NM_000314.6(PTEN):c.289C>T (p.Gln97Ter)	PTEN	10	87933048	not provided	
NM_000314.6(PTEN):c.367C>T (p.His123Tyr)	PTEN	10	87933126	not provided	
NM_000314.6(PTEN):c.385G>A (p.Gly129Arg)	PTEN	10	87933144	not provided	
NM_000314.6(PTEN):c.493-1G>A	PTEN	10	87952117	not provided	
NM_000314.6(PTEN):c.801+1G>A	PTEN	10	87958020	not provided	
NM_005249.4(FOXG1):c.136C>T (p.Gln46Ter)	FOXG1	14	28767415	not provided	
NM_005249.4(FOXG1):c.610C>T (p.Leu204Phe)	FOXG1	14	28767889	not provided	
NM_000891.2(KCNJ2):c.935G>A (p.Arg312His)	KCNJ2	17	70175974	not provided	
NM_001037.4(SCN1B):c.73G>A (p.Asp25Asn)	SCN1B	19	35032560	not provided	
NM_001037.4(SCN1B):c.449-1G>A	SCN1B	19	35039116	not provided	
NM_003165.3(STXBP1):c.874C>T (p.Arg292Cys)	STXBP1	9	127668159	not provided	
NM_003159.2(CDKL5):c.1897C>T (p.Gln633Ter)	CDKL5	X	18604821	not provided	
NM_170707.3(LMNA):c.356+1G>A	LMNA	1	156115275	not provided	
NM_170707.3(LMNA):c.871G>A (p.Glu291Lys)	LMNA	1	156135247	not provided	
NM_170707.3(LMNA):c.1401G>A (p.Trp467Ter)	LMNA	1	156136941	not provided	
NM_170707.3(LMNA):c.1560G>A (p.Trp520Ter)	LMNA	1	156137184	not provided	
NM_001035.2(RYR2):c.689G>A (p.Gly230Asp)	RYR2	1	237388099	not provided	
NM_001035.2(RYR2):c.184C>T (p.Leu62Phe)	RYR2	1	237330893	not provided	
NM_001035.2(RYR2):c.5170G>A (p.Glu1724Lys)	RYR2	1	237614298	not provided	
NM_001035.2(RYR2):c.6940G>A (p.Glu2314Lys)	RYR2	1	237639026	not provided	
NM_001035.2(RYR2):c.11934G>A (p.Met3978Ile)	RYR2	1	237781618	not provided	
NM_001035.2(RYR2):c.12470G>A (p.Arg4157Gln)	RYR2	1	237784182	not provided	
NM_001035.2(RYR2):c.14465G>A (p.Arg4822His)	RYR2	1	237819067	not provided	
NM_001035.2(RYR2):c.14704C>T (p.Pro4902Ser)	RYR2	1	237830578	not provided	
NM_000090.3(COL3A1):c.4072C>T (p.Arg1358Ter)	COL3A1	2	189010708	not provided	
NM_000090.3(COL3A1):c.4087C>T (p.Arg1363Ter)	COL3A1	2	189010723	not provided	
NM_198056.2(SCN5A):c.4035G>A (p.Trp1345Ter)	SCN5A	3	38560357	not provided	
NM_198056.2(SCN5A):c.3994C>T (p.Pro1332Ser)	SCN5A	3	38560398	not provided	
NM_198056.2(SCN5A):c.3575G>A (p.Trp1192Ter)	SCN5A	3	38575388	not provided	
NM_198056.2(SCN5A):c.3553C>T (p.Gln1185Ter)	SCN5A	3	38575410	not provided	
NM_198056.2(SCN5A):c.1891-1G>A	SCN5A	3	38599051	not provided	
NM_198056.2(SCN5A):c.1080G>A (p.Trp360Ter)	SCN5A	3	38606729	not provided	
NM_198056.2(SCN5A):c.664C>T (p.Arg222Ter)	SCN5A	3	38613782	not provided	
NM_198056.2(SCN5A):c.611+1G>A	SCN5A	3	38620842	not provided	
NM_004415.3(DSP):c.151C>T (p.Gln51Ter)	DSP	6	7542066	not provided	
NM_004415.3(DSP):c.273+1G>A	DSP	6	7555821	not provided	
NM_004415.3(DSP):c.3805C>T (p.Arg1269Ter)	DSP	6	7579995	not provided	

NM_004415.3(DSP):c.3961C>T (p.Gln1321Ter)	DSP	6	7580151	not provided
NM_004415.3(DSP):c.4198C>T (p.Arg1400Ter)	DSP	6	7580388	not provided
NM_004415.3(DSP):c.6496C>T (p.Arg2166Ter)	DSP	6	7583758	not provided
NM_004415.3(DSP):c.6850C>T (p.Arg2284Ter)	DSP	6	7584112	not provided
NM_000238.3(KCNH2):c.2104C>T (p.Gln702Ter)	KCNH2	7	150950962	not provided
NM_000238.3(KCNH2):c.2026C>T (p.Gln676Ter)	KCNH2	7	150951040	not provided
NM_000238.3(KCNH2):c.1946-1G>A	KCNH2	7	150951121	not provided
NM_000238.3(KCNH2):c.1943G>A (p.Gly648Asp)	KCNH2	7	150951450	not provided
NM_000238.3(KCNH2):c.1704G>A (p.Trp568Ter)	KCNH2	7	150951689	not provided
NM_000238.3(KCNH2):c.1694C>T (p.Ala565Val)	KCNH2	7	150951699	not provided
NM_000238.3(KCNH2):c.1684C>T (p.His562Tyr)	KCNH2	7	150951709	not provided
NM_000238.3(KCNH2):c.1675C>T (p.Leu559Phe)	KCNH2	7	150951718	not provided
NM_000238.3(KCNH2):c.1193G>A (p.Trp398Ter)	KCNH2	7	150952789	not provided
NM_000238.3(KCNH2):c.1129-1G>A	KCNH2	7	150952854	not provided
NM_000238.3(KCNH2):c.1096C>T (p.Arg366Ter)	KCNH2	7	150957323	not provided
NM_000238.3(KCNH2):c.214C>T (p.Pro72Ser)	KCNH2	7	150974804	not provided
NM_000238.3(KCNH2):c.31C>T (p.Gln11Ter)	KCNH2	7	150977883	not provided
NM_001613.2(ACTA2):c.809G>A (p.Gly270Glu)	ACTA2	10	88938242	not provided
NM_000218.2(KCNQ1):c.343G>A (p.Glu115Lys)	KCNQ1	11	2445441	not provided
NM_000218.2(KCNQ1):c.961C>T (p.Gln321Ter)	KCNQ1	11	2583474	not provided
NM_000218.2(KCNQ1):c.969G>A (p.Trp323Ter)	KCNQ1	11	2583482	not provided
NM_000218.2(KCNQ1):c.974G>A (p.Gly325Glu)	KCNQ1	11	2583487	not provided
NM_000218.2(KCNQ1):c.1176G>A (p.Trp392Ter)	KCNQ1	11	2587617	not provided
NM_000218.2(KCNQ1):c.1336G>A (p.Asp446Asn)	KCNQ1	11	2588797	not provided
NM_000218.2(KCNQ1):c.1660G>A (p.Val554Met)	KCNQ1	11	2776029	not provided
NM_000218.2(KCNQ1):c.1780C>T (p.Arg594Ter)	KCNQ1	11	2778023	not provided
NM_000218.2(KCNQ1):c.1801C>T (p.Gln601Ter)	KCNQ1	11	2847773	not provided
NM_130799.2(MEN1):c.1660C>T (p.Gln554Ter)	MEN1	11	64804507	not provided
NM_130799.2(MEN1):c.1324C>T (p.Gln442Ter)	MEN1	11	64805060	not provided
NM_130799.2(MEN1):c.322C>T (p.Arg108Ter)	MEN1	11	64809788	not provided
NM_000138.4(FBN1):c.7775G>A (p.Cys2592Tyr)	FBN1	15	48420731	not provided
NM_000138.4(FBN1):c.6871G>A (p.Asp2291Asn)	FBN1	15	48430671	not provided
NM_000138.4(FBN1):c.6752G>A (p.Cys2251Tyr)	FBN1	15	48430790	not provided
NM_000138.4(FBN1):c.6496G>A (p.Asp2166Asn)	FBN1	15	48436961	not provided
NM_000138.4(FBN1):c.6425G>A (p.Cys2142Tyr)	FBN1	15	48437032	not provided
NM_000138.4(FBN1):c.6418G>A (p.Gly2140Arg)	FBN1	15	48437039	not provided
NM_000138.4(FBN1):c.6388G>A (p.Glu2130Lys)	FBN1	15	48437069	not provided
NM_000138.4(FBN1):c.5801G>A (p.Cys1934Tyr)	FBN1	15	48445492	not provided
NM_000138.4(FBN1):c.5699G>A (p.Cys1900Tyr)	FBN1	15	48446795	not provided
NM_000138.4(FBN1):c.5513G>A (p.Gly1838Asp)	FBN1	15	48452594	not provided
NM_000138.4(FBN1):c.5015G>A (p.Cys1672Tyr)	FBN1	15	48463949	not provided
NM_000138.4(FBN1):c.4888C>T (p.Gln1630Ter)	FBN1	15	48465622	not provided
NM_000138.4(FBN1):c.4062G>A (p.Trp1354Ter)	FBN1	15	48474553	not provided
NM_000138.4(FBN1):c.3964+1G>A	FBN1	15	48481654	not provided
NM_000138.4(FBN1):c.3712+1G>A	FBN1	15	48485373	not provided

NM_000138.4(FBN1):c.3712G>A (p.Asp1238Asn)	FBN1	15	48485374	not provided	
NM_000138.4(FBN1):c.2926C>T (p.Arg976Cys)	FBN1	15	48490007	not provided	
NM_000138.4(FBN1):c.2806C>T (p.Gln936Ter)	FBN1	15	48492509	not provided	
NM_000138.4(FBN1):c.2671C>T (p.Gln891Ter)	FBN1	15	48495129	not provided	
NM_000138.4(FBN1):c.2539+1G>A	FBN1	15	48495468	not provided	
NM_000138.4(FBN1):c.1583G>A (p.Cys528Tyr)	FBN1	15	48513554	not provided	
NM_000138.4(FBN1):c.1421G>A (p.Cys474Tyr)	FBN1	15	48515434	not provided	
NM_000138.4(FBN1):c.539-1G>A	FBN1	15	48537809	not provided	
NM_000138.4(FBN1):c.164+1G>A	FBN1	15	48644605	not provided	
NM_001943.4(DSG2):c.769C>T (p.Gln257Ter)	DSG2	18	31524526	not provided	
NM_000268.3(NF2):c.810+1G>A	NF2	22	29661340	not provided	
NM_001103.3(ACTN2):c.2527-1G>A	ACTN2	1	236762460	not provided	
NM_001927.3(DES):c.634C>T (p.Arg212Ter)	DES	2	219420150	not provided	
NM_001134363.2(RBM20):c.1156C>T (p.Gln386Ter)	RBM20	10	110781765	not provided	
NM_001134363.2(RBM20):c.1183C>T (p.Gln395Ter)	RBM20	10	110781792	not provided	
NM_001134363.2(RBM20):c.1910G>A (p.Ser637Asn)	RBM20	10	110812307	not provided	
NM_004572.3(PKP2):c.2299+1G>A	PKP2	12	32802402	not provided	
NM_004572.3(PKP2):c.1951C>T (p.Arg651Ter)	PKP2	12	32822487	not provided	
NM_004572.3(PKP2):c.1063C>T (p.Arg355Ter)	PKP2	12	32869034	not provided	
NM_004572.3(PKP2):c.397C>T (p.Gln133Ter)	PKP2	12	32878483	not provided	
NM_004572.3(PKP2):c.369G>A (p.Trp123Ter)	PKP2	12	32878511	not provided	
NM_000116.4(TAZ):c.280C>T (p.Arg94Cys)	TAZ	X	154413248	not provided	
NM_000116.4(TAZ):c.526C>T (p.His176Tyr)	TAZ	X	154419608	not provided	
NM_000116.4(TAZ):c.582G>A (p.Trp194Ter)	TAZ	X	154419745	not provided	
NM_001256850.1(TTN):c.96304C>T (p.Arg32102Ter)	TTN	2	178535388	not provided	
NM_001256850.1(TTN):c.91774C>T (p.Arg30592Ter)	TTN	2	178543276	not provided	
NM_001256850.1(TTN):c.82828C>T (p.Arg27610Ter)	TTN	2	178557511	not provided	
NM_001256850.1(TTN):c.81193C>T (p.Arg27065Ter)	TTN	2	178560016	not provided	
NM_001256850.1(TTN):c.79974G>A (p.Trp26658Ter)	TTN	2	178561235	not provided	
NM_001256850.1(TTN):c.72514C>T (p.Gln24172Ter)	TTN	2	178568695	not provided	
NM_001256850.1(TTN):c.70405C>T (p.Arg23469Ter)	TTN	2	178570804	not provided	
NM_001256850.1(TTN):c.70327C>T (p.Arg23443Ter)	TTN	2	178570882	not provided	
NM_001256850.1(TTN):c.68923C>T (p.Arg22975Ter)	TTN	2	178572286	not provided	
NM_001256850.1(TTN):c.64327C>T (p.Arg21443Ter)	TTN	2	178577085	not provided	
NM_001256850.1(TTN):c.60076C>T (p.Arg20026Ter)	TTN	2	178584552	not provided	
NM_001256850.1(TTN):c.58702C>T (p.Arg19568Ter)	TTN	2	178587684	not provided	
NM_001256850.1(TTN):c.55198C>T (p.Gln18400Ter)	TTN	2	178591698	not provided	
NM_001256850.1(TTN):c.49637G>A (p.Trp16546Ter)	TTN	2	178604127	not provided	
NM_001256850.1(TTN):c.48432G>A (p.Trp16144Ter)	TTN	2	178607247	not provided	
NM_001256850.1(TTN):c.48076C>T (p.Gln16026Ter)	TTN	2	178607788	not provided	
NM_001256850.1(TTN):c.47550G>A (p.Trp15850Ter)	TTN	2	178608410	not provided	
NM_001256850.1(TTN):c.45160C>T (p.Arg15054Ter)	TTN	2	178612442	not provided	
NM_001256850.1(TTN):c.43383G>A (p.Trp14461Ter)	TTN	2	178616485	not provided	
NM_001256850.1(TTN):c.43360C>T (p.Arg14454Ter)	TTN	2	178616508	not provided	
NM_001256850.1(TTN):c.42556C>T (p.Gln14186Ter)	TTN	2	178617872	not provided	

NM_145207.2(SPATA5):c.251G>A (p.Arg84Gln)	SPATA5	4	122927721	not provided	
NM_015506.2(MMACHC):c.217C>T (p.Arg73Ter)	MMACHC	1	45507491	not provided	
NM_015506.2(MMACHC):c.440G>A (p.Gly147Asp)	MMACHC	1	45508806	not provided	
NM_020166.4(MCCC1):c.137G>A (p.Gly46Glu)	MCCC1	3	183092545	not provided	
NM_000387.5(SLC25A20):c.10C>T (p.Gln4Ter)	SLC25A20	3	48898785	not provided	
NM_004453.3(ETFDH):c.1073G>A (p.Arg358Lys)	ETFDH	4	158699087	not provided	
NM_004453.3(ETFDH):c.1809G>A (p.Trp603Ter)	ETFDH	4	158708482	not provided	
NM_003060.3(SLC22A5):c.42G>A (p.Trp14Ter)	SLC22A5	5	132370014	not provided	
NM_000255.3(MUT):c.682C>T (p.Arg228Ter)	MUT	6	49457762	not provided	
NM_000048.3(ASL):c.649C>T (p.Arg217Ter)	ASL	7	66087380	not provided	
NM_000050.4(ASS1):c.1088G>A (p.Arg363Gln)	ASS1	9	130494984	not provided	
NM_000017.3(ACADS):c.988C>T (p.Arg330Cys)	ACADS	12	120738874	not provided	
NM_174917.4(ACSF3):c.589G>A (p.Ala197Thr)	ACSF3	16	89101270	not provided	
NM_000018.3(ACADVL):c.881G>A (p.Gly294Glu)	ACADVL	17	7222669	not provided	
NM_000018.3(ACADVL):c.1096C>T (p.Arg366Cys)	ACADVL	17	7223151	not provided	
NM_000018.3(ACADVL):c.1316G>A (p.Gly439Asp)	ACADVL	17	7223859	not provided	
NM_000310.3(PPT1):c.362+5G>A	PPT1	1	40092040	not provided	
NM_000310.3(PPT1):c.234+1G>A	PPT1	1	40092397	not provided	
NM_006516.2(SLC2A1):c.667C>T (p.Arg223Trp)	SLC2A1	1	42929885	not provided	
NM_006516.2(SLC2A1):c.18+1G>A	SLC2A1	1	42958633	not provided	
NM_021007.2(SCN2A):c.823C>T (p.Arg275Ter)	SCN2A	2	165310448	not provided	
NM_021007.2(SCN2A):c.2809C>T (p.Arg937Cys)	SCN2A	2	165344801	not provided	
NM_021007.2(SCN2A):c.2995G>A (p.Glu999Lys)	SCN2A	2	165354267	not provided	
NM_021007.2(SCN2A):c.3944G>A (p.Arg1315Lys)	SCN2A	2	165373319	not provided	
NM_021007.2(SCN2A):c.4877G>A (p.Arg1626Gln)	SCN2A	2	165388683	not provided	
NM_021007.2(SCN2A):c.4886G>A (p.Arg1629His)	SCN2A	2	165388692	not provided	
NM_001165963.1(SCN1A):c.5710C>T (p.Gln1904Ter)	SCN1A	2	165991565	not provided	
NM_001165963.1(SCN1A):c.5435G>A (p.Trp1812Ter)	SCN1A	2	165991840	not provided	
NM_001165963.1(SCN1A):c.4931G>A (p.Gly1644Asp)	SCN1A	2	165992344	not provided	
NM_001165963.1(SCN1A):c.4285G>A (p.Ala1429Thr)	SCN1A	2	165999776	not provided	
NM_001165963.1(SCN1A):c.3607C>T (p.Gln1203Ter)	SCN1A	2	166013842	not provided	
NM_001165963.1(SCN1A):c.2782C>T (p.Gln928Ter)	SCN1A	2	166037940	not provided	
NM_001165963.1(SCN1A):c.1662G>A (p.Gln554=)	SCN1A	2	166045043	not provided	
NM_001165963.1(SCN1A):c.1348C>T (p.Gln450Ter)	SCN1A	2	166046799	not provided	
NM_001165963.1(SCN1A):c.842C>T (p.Pro281Leu)	SCN1A	2	166051841	not provided	
NM_001165963.1(SCN1A):c.603-1G>A	SCN1A	2	166052944	not provided	
NM_001165963.1(SCN1A):c.314C>T (p.Thr105Ile)	SCN1A	2	166058639	not provided	
NM_001165963.1(SCN1A):c.265-1G>A	SCN1A	2	166058689	not provided	
NM_001165963.1(SCN1A):c.82C>T (p.Arg28Cys)	SCN1A	2	166073540	not provided	
NM_152778.2(MFSD8):c.599G>A (p.Trp200Ter)	MFSD8	4	127939952	not provided	
NM_001182.4(ALDH7A1):c.1004G>A (p.Arg335Gln)	ALDH7A1	5	126559244	not provided	
NM_000816.3(GABRG2):c.316G>A (p.Ala106Thr)	GABRG2	5	162095551	not provided	
NM_000816.3(GABRG2):c.770-1G>A	GABRG2	5	162142163	not provided	
NM_000816.3(GABRG2):c.1336C>T (p.Arg446Trp)	GABRG2	5	162153300	not provided	
NM_002397.4(MEF2C):c.43C>T (p.Arg15Cys)	MEF2C	5	88823746	not provided	

NM_018100.3(EFHC1):c.1612C>T (p.Arg538Ter)	EFHC1	6	52479759	not provided	
NM_014141.5(CNTNAP2):c.1447C>T (p.Arg483Ter)	CNTNAP2	7	147300239	not provided	
NM_004519.3(KCNQ3):c.679C>T (p.Arg227Ter)	KCNQ3	8	132180255	not provided	
NM_003165.3(STXBP1):c.87+1G>A	STXBP1	9	127651653	not provided	
NM_003165.3(STXBP1):c.569G>A (p.Arg190Gln)	STXBP1	9	127663344	not provided	
NM_003165.3(STXBP1):c.1061G>A (p.Cys354Tyr)	STXBP1	9	127673212	not provided	
NM_003165.3(STXBP1):c.1359+1G>A	STXBP1	9	127676754	not provided	
NM_003165.3(STXBP1):c.1672C>T (p.Gln558Ter)	STXBP1	9	127682530	not provided	
NM_003165.3(STXBP1):c.1702+1G>A	STXBP1	9	127682561	not provided	
NM_001909.4(CTSD):c.299C>T (p.Ser100Phe)	CTSD	11	1759569	not provided	
NM_000391.3(TPP1):c.509-1G>A	TPP1	11	6617154	not provided	
NM_000391.3(TPP1):c.379C>T (p.Arg127Ter)	TPP1	11	6617627	not provided	
NM_000391.3(TPP1):c.196C>T (p.Gln66Ter)	TPP1	11	6618809	not provided	
NM_014191.3(SCN8A):c.4949C>T (p.Ala1650Val)	SCN8A	12	51806435	not provided	
NM_006493.2(CLN5):c.595C>T (p.Arg199Ter)	CLN5	13	76996010	not provided	
NM_005249.4(FOXG1):c.217C>T (p.Gln73Ter)	FOXG1	14	28767496	not provided	
NM_005249.4(FOXG1):c.586C>T (p.Gln196Ter)	FOXG1	14	28767865	not provided	
NM_005249.4(FOXG1):c.667C>T (p.Gln223Ter)	FOXG1	14	28767946	not provided	
NM_005249.4(FOXG1):c.671G>A (p.Gly224Asp)	FOXG1	14	28767950	not provided	
NM_001482.2(GATM):c.778C>T (p.Arg260Ter)	GATM	15	45366406	not provided	
NM_017882.2(CLN6):c.665+1G>A	CLN6	15	68209636	not provided	
NM_017882.2(CLN6):c.297+1G>A	CLN6	15	68214289	not provided	
NM_002693.2(POLG):c.3287G>A (p.Arg1096His)	POLG	15	89318736	not provided	
NM_002693.2(POLG):c.3286C>T (p.Arg1096Cys)	POLG	15	89318737	not provided	
NM_002693.2(POLG):c.3139C>T (p.Arg1047Trp)	POLG	15	89319065	not provided	
NM_002693.2(POLG):c.2558G>A (p.Arg853Gln)	POLG	15	89321776	not provided	
NM_002693.2(POLG):c.2420G>A (p.Arg807His)	POLG	15	89322748	not provided	
NM_002693.2(POLG):c.1716G>A (p.Trp572Ter)	POLG	15	89325683	not provided	
NM_000548.4(TSC2):c.2639+1G>A	TSC2	16	2073893	not provided	
NM_000548.4(TSC2):c.3180G>A (p.Trp1060Ter)	TSC2	16	2079324	not provided	
NM_000548.4(TSC2):c.3624G>A (p.Trp1208Ter)	TSC2	16	2081608	not provided	
NM_001042432.1(CLN3):c.988G>A (p.Val330Ile)	CLN3	16	28482173	not provided	
NM_001042432.1(CLN3):c.949C>T (p.Gln317Ter)	CLN3	16	28482340	not provided	
NM_001193466.1(KANSL1):c.2203+1G>A	KANSL1	17	46039701	not provided	
NM_018129.3(PNPO):c.673C>T (p.Arg225Cys)	PNPO	17	47946669	not provided	
NM_001083962.1(TCF4):c.1570C>T (p.Gln524Ter)	TCF4	18	55232588	not provided	
NM_000156.5(GAMT):c.491G>A (p.Gly164Asp)	GAMT	19	1398995	not provided	
NM_007254.3(PNKP):c.992G>A (p.Trp331Ter)	PNKP	19	49862408	not provided	
NM_007254.3(PNKP):c.610C>T (p.Arg204Ter)	PNKP	19	49864205	not provided	
NM_007254.3(PNKP):c.490C>T (p.Gln164Ter)	PNKP	19	49865135	not provided	
NM_172107.3(KCNQ2):c.1632-1G>A	KCNQ2	20	63413582	not provided	
NM_172107.3(KCNQ2):c.1009G>A (p.Ala337Thr)	KCNQ2	20	63438639	not provided	
NM_172107.3(KCNQ2):c.1004C>T (p.Pro335Leu)	KCNQ2	20	63438644	not provided	
NM_172107.3(KCNQ2):c.844G>A (p.Asp282Asn)	KCNQ2	20	63439681	not provided	
NM_172107.3(KCNQ2):c.809G>A (p.Trp270Ter)	KCNQ2	20	63442413	not provided	

NM_172107.3(KCNQ2):c.740C>T (p.Ser247Leu)	KCNQ2	20	63442482	not provided	
NM_172107.3(KCNQ2):c.593G>A (p.Arg198Gln)	KCNQ2	20	63444756	not provided	
NM_000026.3(ADSL):c.5C>T (p.Ala2Val)	ADSL	22	40346563	not provided	
NM_000026.3(ADSL):c.568C>T (p.Arg190Ter)	ADSL	22	40358949	not provided	
NM_000026.3(ADSL):c.853C>T (p.Gln285Ter)	ADSL	22	40361333	not provided	
NM_000026.3(ADSL):c.1009C>T (p.Arg337Ter)	ADSL	22	40361634	not provided	
NM_000026.3(ADSL):c.1187G>A (p.Arg396His)	ADSL	22	40364361	not provided	
NM_000026.3(ADSL):c.1276C>T (p.Arg426Cys)	ADSL	22	40364964	not provided	
NM_001184880.1(PCDH19):c.2656C>T (p.Arg886Ter)	PCDH19	X	100350665	not provided	
NM_001184880.1(PCDH19):c.1114C>T (p.Arg372Trp)	PCDH19	X	100407484	not provided	
NM_001184880.1(PCDH19):c.1031C>T (p.Pro344Leu)	PCDH19	X	100407567	not provided	
NM_001184880.1(PCDH19):c.619C>T (p.Arg207Ter)	PCDH19	X	100407979	not provided	
NM_006359.2(SLC9A6):c.1631+1G>A	SLC9A6	X	136033494	not provided	
NM_133499.2(SYN1):c.1264C>T (p.Arg422Ter)	SYN1	X	47575169	not provided	
NM_001193416.2(DDX3X):c.1463G>A (p.Arg488His)	DDX3X	X	41346376	not provided	
NM_003242.5(TGFBR2):c.1276G>A (p.Ala426Thr)	TGFBR2	3	30674126	not provided	
NM_003242.5(TGFBR2):c.1279C>T (p.Pro427Ser)	TGFBR2	3	30674129	not provided	
NM_003242.5(TGFBR2):c.1564G>A (p.Asp522Asn)	TGFBR2	3	30691459	not provided	
NM_001999.3(FBN2):c.3593G>A (p.Cys1198Tyr)	FBN2	5	128338002	not provided	
NM_002890.2(RASA1):c.2131C>T (p.Arg711Ter)	RASA1	5	87376512	not provided	
NM_002890.2(RASA1):c.2690+1G>A	RASA1	5	87380596	not provided	
NM_000118.3(ENG):c.1428+1G>A	ENG	9	127818715	not provided	
NM_000118.3(ENG):c.1272+1G>A	ENG	9	127819899	not provided	
NM_000118.3(ENG):c.562C>T (p.Gln188Ter)	ENG	9	127825822	not provided	
NM_000118.3(ENG):c.446G>A (p.Trp149Ter)	ENG	9	127826587	not provided	
NM_000118.3(ENG):c.247C>T (p.Gln83Ter)	ENG	9	127829800	not provided	
NM_000093.4(COL5A1):c.2734C>T (p.Arg912Ter)	COL5A1	9	134795115	not provided	
NM_000093.4(COL5A1):c.4282C>T (p.Gln1428Ter)	COL5A1	9	134818707	not provided	
NM_000093.4(COL5A1):c.4474G>A (p.Gly1492Ser)	COL5A1	9	134820143	not provided	
NM_004612.3(TGFBR1):c.973+1G>A	TGFBR1	9	99142704	not provided	
NM_000192.3(TBX5):c.1126C>T (p.Gln376Ter)	TBX5	12	114355963	not provided	
NM_000192.3(TBX5):c.755G>A (p.Ser252Asn)	TBX5	12	114385476	not provided	
NM_000192.3(TBX5):c.142C>T (p.Gln48Ter)	TBX5	12	114403757	not provided	
NM_000020.2(ACVRL1):c.152G>A (p.Cys51Tyr)	ACVRL1	12	51913189	not provided	
NM_000020.2(ACVRL1):c.200G>A (p.Arg67Gln)	ACVRL1	12	51913237	not provided	
NM_000020.2(ACVRL1):c.269G>A (p.Cys90Tyr)	ACVRL1	12	51913306	not provided	
NM_000020.2(ACVRL1):c.986G>A (p.Arg329His)	ACVRL1	12	51915438	not provided	
NM_000020.2(ACVRL1):c.1377+1G>A	ACVRL1	12	51919116	not provided	
NM_000214.2(JAG1):c.2230C>T (p.Arg744Ter)	JAG1	20	10644977	not provided	
NM_000214.2(JAG1):c.910C>T (p.Gln304Ter)	JAG1	20	10652227	not provided	
NM_000214.2(JAG1):c.544C>T (p.Gln182Ter)	JAG1	20	10658618	not provided	
NM_000214.2(JAG1):c.439+1G>A	JAG1	20	10663962	not provided	
NM_000071.2(CBS):c.1280C>T (p.Pro427Leu)	CBS	21	43058912	not provided	
NM_000071.2(CBS):c.1135C>T (p.Arg379Trp)	CBS	21	43060451	not provided	
NM_000071.2(CBS):c.700G>A (p.Asp234Asn)	CBS	21	43065239	not provided	

NM_000071.2(CBS):c.253G>A (p.Gly85Arg)	CBS	21	43068572	not provided	
NM_000143.3(FH):c.736C>T (p.Gln246Ter)	FH	1	241508605	not provided	
NM_000143.3(FH):c.7C>T (p.Arg3Ter)	FH	1	241519716	not provided	
NM_002437.4(MPV17):c.370C>T (p.Gln124Ter)	MPV17	2	27312499	not provided	
NM_080916.2(DGUOK):c.591G>A (p.Gln197=)	DGUOK	2	73950732	not provided	
NM_024996.5(GFM1):c.3G>A (p.Met1Ile)	GFM1	3	158644637	not provided	
NM_024996.5(GFM1):c.688G>A (p.Gly230Ser)	GFM1	3	158649156	not provided	
NM_024996.5(GFM1):c.700C>T (p.Arg234Ter)	GFM1	3	158652106	not provided	
NM_024996.5(GFM1):c.2011C>T (p.Arg671Cys)	GFM1	3	158690264	not provided	
NM_015560.2(OPA1):c.871-1G>A	OPA1	3	193637951	not provided	
NM_006005.3(WFS1):c.631G>A (p.Asp211Asn)	WFS1	4	6291367	not provided	
NM_006005.3(WFS1):c.2020G>A (p.Gly674Arg)	WFS1	4	6301815	not provided	
NM_015697.7(COQ2):c.950C>T (p.Thr317Met)	COQ2	4	83267737	not provided	
NM_012208.3(HARS2):c.633+1G>A	HARS2	5	140695846	not provided	
NM_032861.3(SERAC1):c.1126C>T (p.Gln376Ter)	SERAC1	6	158120465	not provided	
NM_020745.3(AARS2):c.595C>T (p.Arg199Cys)	AARS2	6	44311148	not provided	
NM_020320.4(RARS2):c.943C>T (p.Arg315Ter)	RARS2	6	87524588	not provided	
NM_022445.3(TPK1):c.185+1G>A	TPK1	7	144682908	not provided	
NM_015713.4(RRM2B):c.599G>A (p.Gly200Glu)	RRM2B	8	102218899	not provided	
NM_015713.4(RRM2B):c.520C>T (p.Arg174Ter)	RRM2B	8	102224076	not provided	
NM_175073.2(APTX):c.667C>T (p.Leu223Phe)	APTX	9	32984734	not provided	
NM_021830.4(TWNK):c.967C>T (p.Arg323Ter)	TWNK	10	100989177	not provided	
NM_017547.3(FOXRED1):c.86-1G>A	FOXRED1	11	126271436	not provided	
NM_007103.3(NDUFV1):c.349G>A (p.Ala117Thr)	NDUFV1	11	67609474	not provided	
NM_007103.3(NDUFV1):c.365C>T (p.Pro122Leu)	NDUFV1	11	67609490	not provided	
NM_007103.3(NDUFV1):c.617G>A (p.Cys206Tyr)	NDUFV1	11	67610487	not provided	
NM_021957.3(GYS2):c.574C>T (p.Arg192Ter)	GYS2	12	21574248	not provided	
NM_021957.3(GYS2):c.547C>T (p.Gln183Ter)	GYS2	12	21574275	not provided	
NM_005262.2(GFER):c.373C>T (p.Gln125Ter)	GFER	16	1984861	not provided	
NM_017775.3(TTC19):c.194G>A (p.Trp65Ter)	TTC19	17	16000127	not provided	
NM_000151.3(G6PC):c.562G>A (p.Gly188Ser)	G6PC	17	42909418	not provided	
NM_017827.3(SARS2):c.589+1G>A	SARS2	19	38921391	not provided	
NM_017827.3(SARS2):c.30G>A (p.Trp10Ter)	SARS2	19	38930707	not provided	
NM_018006.4(TRMU):c.718C>T (p.Arg240Ter)	TRMU	22	46352276	not provided	
NM_000284.3(PDHA1):c.292-1G>A	PDHA1	X	19351280	not provided	
NM_000284.3(PDHA1):c.506C>T (p.Ala169Val)	PDHA1	X	19353169	not provided	
NM_000284.3(PDHA1):c.910C>T (p.Arg304Ter)	PDHA1	X	19358926	not provided	
NM_000032.4(ALAS2):c.1355G>A (p.Arg452His)	ALAS2	X	55014829	not provided	
NM_000032.4(ALAS2):c.606G>A (p.Met202Ile)	ALAS2	X	55021084	not provided	
NM_003990.4(PAX2):c.187G>A (p.Gly63Ser)	PAX2	10	100749889	not provided	
NM_001287174.1(ABCC8):c.683G>A (p.Gly228Asp)	ABCC8	11	17461722	not provided	
NM_006245.3(PPP2R5D):c.589G>A (p.Glu197Lys)	PPP2R5D	6	43007262	not provided	
NM_006245.3(PPP2R5D):c.1258G>A (p.Glu420Lys)	PPP2R5D	6	43009328	not provided	
NM_001164144.2(CHAMP1):c.1544G>A (p.Trp515Ter)	CHAMP1	13	114325386	not provided	
NM_001164144.2(CHAMP1):c.1945C>T (p.Gln649Ter)	CHAMP1	13	114325787	not provided	

NM_032436.3(CHAMP1):c.1969C>T (p.Gln657Ter)	CHAMP1	13	114325811	not provided	
NM_000038.5(APC):c.221-1G>A	APC	5	112767188	not provided	
NM_000038.5(APC):c.487C>T (p.Gln163Ter)	APC	5	112775693	not provided	
NM_000038.5(APC):c.541C>T (p.Gln181Ter)	APC	5	112780799	not provided	
NM_000038.5(APC):c.1297C>T (p.Gln433Ter)	APC	5	112819329	not provided	
NM_000038.5(APC):c.1312+1G>A	APC	5	112819345	not provided	
NM_000038.5(APC):c.1959-1G>A	APC	5	112837552	not provided	
NM_000038.5(APC):c.3688C>T (p.Gln1230Ter)	APC	5	112839282	not provided	
NM_000038.5(APC):c.4645C>T (p.Gln1549Ter)	APC	5	112840239	not provided	
NM_000147.4(FUCA1):c.1082G>A (p.Trp361Ter)	FUCA1	1	23848727	not provided	
NM_001904.3(CTNNB1):c.1867C>T (p.Gln623Ter)	CTNNB1	3	41236412	not provided	
NM_001256214.1(ATP1A3):c.1006C>T (p.Pro336Ser)	ATP1A3	19	41984944	not provided	
NM_006734.3(HIVEP2):c.2905C>T (p.Gln969Ter)	HIVEP2	6	142771834	not provided	
NM_000369.2(TSHR):c.1349G>A (p.Arg450His)	TSHR	14	81143407	not provided	
NM_001012614.1(CTBP1):c.991C>T (p.Arg331Trp)	CTBP1	4	1213028	not provided	
NM_002465.3(MYBPC1):c.742G>A (p.Glu248Lys)	MYBPC1	12	101642495	not provided	
NM_018136.4(ASPM):c.4422G>A (p.Trp1474Ter)	ASPM	1	197104829	not provided	
NM_004895.4(NLRP3):c.1711G>A (p.Gly571Arg)	NLRP3	1	247425154	not provided	
NM_004321.7(KIF1A):c.506G>A (p.Arg169Lys)	KIF1A	2	240786437	not provided	
NM_000535.6(PMS2):c.7C>T (p.Arg3Ter)	PMS2	7	6009013	not provided	
NM_000431.3(MVK):c.643C>T (p.Arg215Ter)	MVK	12	109586765	not provided	
NM_000834.3(GRIN2B):c.3332G>A (p.Arg1111His)	GRIN2B	12	13563906	not provided	
NM_000059.3(BRCA2):c.4651C>T (p.Gln1551Ter)	BRCA2	13	32339006	not provided	
NM_000243.2(MEFV):c.1759+1G>A	MEFV	16	3244253	not provided	
NM_000214.2(JAG1):c.2566C>T (p.Gln856Ter)	JAG1	20	10642494	not provided	
NM_000214.2(JAG1):c.871C>T (p.Gln291Ter)	JAG1	20	10652483	not provided	
NM_000214.2(JAG1):c.694+1G>A	JAG1	20	10658467	not provided	
NM_000214.2(JAG1):c.81+1G>A	JAG1	20	10673449	not provided	
NM_001456.3(FLNA):c.7231C>T (p.Arg2411Ter)	FLNA	X	154350109	not provided	
NM_002218.2(KCNQ1):c.528G>A (p.Trp176Ter)	KCNQ1	11	2570678	not provided	
NM_000411.6(HLCS):c.2005C>T (p.His669Tyr)	HLCS	21	36756546	not provided	
NM_001080463.1(DYNC2H1):c.11671-1G>A	DYNC2H1	11	103316544	not provided	
NM_000342.3(SLC4A1):c.1825G>A (p.Gly609Arg)	SLC4A1	17	44255272	not provided	
NM_025074.6(FRAS1):c.10948C>T (p.Gln3650Ter)	FRAS1	4	78534471	not provided	
NM_001271208.1(NEB):c.19944G>A (p.Ser6648=)	NEB	2	151551738	not provided	
NM_001130144.2(LTBP3):c.3364C>T (p.Gln1122Ter)	LTBP3	11	65540034	not provided	
NM_007055.3(POLR3A):c.2617C>T (p.Arg873Ter)	POLR3A	10	77993367	not provided	
NM_170740.1(ALDH5A1):c.842G>A (p.Gly281Glu)	ALDH5A1	6	24515243	not provided	
NM_003124.4(SPR):c.655C>T (p.Arg219Ter)	SPR	2	72891406	not provided	
NM_000159.3(GCDH):c.1156C>T (p.Arg386Ter)	GCDH	19	12897776	not provided	
NM_000352.4(ABCC8):c.3640C>T (p.Arg1214Trp)	ABCC8	11	17402671	not provided	
NM_006015.4(ARID1A):c.1207C>T (p.Gln403Ter)	ARID1A	1	26729720	not provided	
NM_000497.3(CYP11B1):c.1331G>A (p.Gly444Asp)	CYP11B1	8	142875024	not provided	
NM_001161766.1(AHCY):c.209C>T (p.Pro70Leu)	AHCY	20	34294083	not provided	
NM_006929.4(SKIV2L):c.1120C>T (p.Arg374Ter)	SKIV2L	6	31962494	not provided	

NM_000016.5(ACADM):c.849+1G>A	ACADM	1	75749560	not provided	
NM_002180.2(IGHMBP2):c.1808G>A (p.Arg603His)	IGHMBP2	11	68936288	not provided	
NM_000426.3(LAMA2):c.1303C>T (p.Arg435Ter)	LAMA2	6	129165672	not provided	
NM_000495.4(COL4A5):c.1339+1G>A	COL4A5	X	108591232	not provided	
NM_024422.4(DSC2):c.1660C>T (p.Gln554Ter)	DSC2	18	31079850	not provided	
NM_001972.3(ELANE):c.597+1G>A	ELANE	19	855795	not provided	
NM_001008537.2(KIAA2022):c.652C>T (p.Arg218Ter)	KIAA2022	X	74743905	not provided	
NM_001008537.2(KIAA2022):c.937C>T (p.Arg313Ter)	KIAA2022	X	74743620	not provided	
NM_014874.3(MFN2):c.1198C>T (p.Arg400Ter)	MFN2	1	12004029	not provided	
NM_170707.3(LMNA):c.513+1G>A	LMNA	1	156130774	not provided	
NM_001012331.1(NTRK1):c.526C>T (p.Gln176Ter)	NTRK1	1	156868201	not provided	
NM_002977.3(SCN9A):c.4462C>T (p.Arg1488Ter)	SCN9A	2	166204368	not provided	
NM_000465.3(BARD1):c.1811-1G>A	BARD1	2	214745160	not provided	
NM_004321.7(KIF1A):c.760C>T (p.Arg254Trp)	KIF1A	2	240783777	not provided	
NM_014845.5(FIG4):c.1207C>T (p.Gln403Ter)	FIG4	6	109760319	not provided	
NM_014845.5(FIG4):c.2459+1G>A	FIG4	6	109792665	not provided	
NM_018972.2(GDAP1):c.769C>T (p.Arg257Ter)	GDAP1	8	74364059	not provided	
NM_000051.3(ATM):c.5414G>A (p.Trp1805Ter)	ATM	11	108302947	not provided	
NM_000051.3(ATM):c.5573G>A (p.Trp1858Ter)	ATM	11	108304751	not provided	
NM_000051.3(ATM):c.5623C>T (p.Arg1875Ter)	ATM	11	108304801	not provided	
NM_000051.3(ATM):c.7449G>A (p.Trp2483Ter)	ATM	11	108330355	not provided	
NM_030962.3(SBF2):c.4315C>T (p.Gln1439Ter)	SBF2	11	9808128	not provided	
NM_000834.3(GRIN2B):c.2539C>T (p.Arg847Ter)	GRIN2B	12	13567084	not provided	
NM_001376.4(DYNC1H1):c.5884C>T (p.Arg1962Cys)	DYNC1H1	14	102008244	not provided	
NM_001376.4(DYNC1H1):c.9092C>T (p.Thr3031Met)	DYNC1H1	14	102027662	not provided	
NM_001376.4(DYNC1H1):c.10420C>T (p.Arg3474Trp)	DYNC1H1	14	102033982	not provided	
NM_024675.3(PALB2):c.3492G>A (p.Trp1164Ter)	PALB2	16	23603528	not provided	
NM_000304.3(PMP22):c.449G>A (p.Gly150Asp)	PMP22	17	15230951	not provided	
NM_007294.3(BRCA1):c.135-1G>A	BRCA1	17	43106534	not provided	
NM_001005360.2(DNM2):c.1678G>A (p.Glu560Lys)	DNM2	19	10819986	not provided	
NM_181882.2(PRX):c.1174C>T (p.Arg392Ter)	PRX	19	40397178	not provided	
NM_001972.3(ELANE):c.597+5G>A	ELANE	19	855799	not provided	
NM_000169.2(GLA):c.1081G>A (p.Gly361Arg)	GLA	X	101398018	not provided	
NM_000169.2(GLA):c.707G>A (p.Trp236Ter)	GLA	X	101398879	not provided	
NM_000166.5(GJB1):c.112G>A (p.Val38Met)	GJB1	X	71223819	not provided	
NM_000166.5(GJB1):c.231G>A (p.Trp77Ter)	GJB1	X	71223938	not provided	
NM_000166.5(GJB1):c.541G>A (p.Val181Met)	GJB1	X	71224248	not provided	
NM_000166.5(GJB1):c.643C>T (p.Arg215Trp)	GJB1	X	71224350	not provided	
NM_144672.3(OTOA):c.755G>A (p.Trp252Ter)	OTOA	16	21697790	not provided	
NM_001429.3(EP300):c.3163C>T (p.Arg1055Ter)	EP300	22	41155015	not provided	
NM_001854.3(COL11A1):c.1685G>A (p.Gly562Asp)	COL11A1	1	103006314	not provided	
NM_002016.1(FLG):c.10225C>T (p.Arg3409Ter)	FLG	1	152304661	not provided	
NM_002016.1(FLG):c.2476C>T (p.Arg826Ter)	FLG	1	152312410	not provided	
NM_020699.3(GATAD2B):c.1432C>T (p.Arg478Ter)	GATAD2B	1	153812120	not provided	
NM_006147.3(IRF6):c.668-1G>A	IRF6	1	209790888	not provided	

NM_000702.3(ATP1A2):c.2500C>T (p.Arg834Ter)	ATP1A2	1	160136307	not provided	
NM_019026.4(TMCO1):c.340C>T (p.Arg114Ter)	TMCO1	1	165759546	not provided	
NM_000721.3(CACNA1E):c.10546>A (p.Gly352Arg)	CACNA1E	1	181651440	not provided	
NM_000228.2(LAMB3):c.1132+5G>A	LAMB3	1	209629732	not provided	
NM_006147.3(IRF6):c.859C>T (p.Gln287Ter)	IRF6	1	209790696	not provided	
NM_006147.3(IRF6):c.576G>A (p.Trp192Ter)	IRF6	1	209792360	not provided	
NM_006147.3(IRF6):c.226C>T (p.Pro76Ser)	IRF6	1	209796501	not provided	
NM_206933.2(USH2A):c.5836C>T (p.Arg1946Ter)	USH2A	1	216072910	not provided	
NM_005529.6(HSPG2):c.7006+1G>A	HSPG2	1	21851790	not provided	
NM_001100.3(ACTA1):c.430C>T (p.Leu144Phe)	ACTA1	1	229432580	not provided	
NM_006516.2(SLC2A1):c.635G>A (p.Arg212His)	SLC2A1	1	42929917	not provided	
NM_005373.2(MPL):c.317C>T (p.Pro106Leu)	MPL	1	43338646	not provided	
NM_017739.3(POMGNT1):c.636C>T (p.Phe212=)	POMGNT1	1	46194860	not provided	
NM_005595.4(NFIA):c.361C>T (p.Arg121Cys)	NFIA	1	61088482	not provided	
NM_000350.2(ABCA4):c.1906C>T (p.Gln636Ter)	ABCA4	1	94062608	not provided	
NM_022787.3(NMNAT1):c.507G>A (p.Trp169Ter)	NMNAT1	1	9982368	not provided	
NM_022336.3(EDAR):c.1121G>A (p.Trp374Ter)	EDAR	2	108897133	not provided	
NM_022336.3(EDAR):c.1073G>A (p.Arg358Gln)	EDAR	2	108897181	not provided	
NM_022336.3(EDAR):c.212G>A (p.Cys71Tyr)	EDAR	2	108929342	not provided	
NM_015702.2(MMADHC):c.472C>T (p.Arg158Ter)	MMADHC	2	149576443	not provided	
NM_001271208.1(NEB):c.12018+1G>A	NEB	2	151610515	not provided	
NM_021007.2(SCN2A):c.4308+1G>A	SCN2A	2	165377651	not provided	
NM_001165963.1(SCN1A):c.3857G>A (p.Trp1286Ter)	SCN1A	2	166012131	not provided	
NM_001165963.1(SCN1A):c.1702C>T (p.Arg568Ter)	SCN1A	2	166044010	not provided	
NM_001256850.1(TTN):c.44490G>A (p.Trp14830Ter)	TTN	2	178613870	not provided	
NM_000393.4(COL5A2):c.2627G>A (p.Gly876Glu)	COL5A2	2	189052945	not provided	
NM_001204.6(BMPR2):c.1789C>T (p.Arg597Ter)	BMPR2	2	202555454	not provided	
NM_173076.2(ABCA12):c.3889C>T (p.Arg1297Ter)	ABCA12	2	214987734	not provided	
NM_173076.2(ABCA12):c.2140C>T (p.Arg714Ter)	ABCA12	2	215011631	not provided	
NM_173076.2(ABCA12):c.1300C>T (p.Arg434Ter)	ABCA12	2	215019784	not provided	
NM_173076.2(ABCA12):c.859C>T (p.Arg287Ter)	ABCA12	2	215045850	not provided	
NM_173076.2(ABCA12):c.178C>T (p.Arg60Ter)	ABCA12	2	215064205	not provided	
NM_025216.2(WNT10A):c.742C>T (p.Arg248Ter)	WNT10A	2	218890349	not provided	
NM_025243.3(SLC19A3):c.1314+1G>A	SLC19A3	2	227688165	not provided	
NM_014946.3(SPAST):c.508C>T (p.Gln170Ter)	SPAST	2	32089527	not provided	
NM_001298.2(CNGA3):c.1688G>A (p.Arg563His)	CNGA3	2	98396858	not provided	
NM_000532.4(PCCB):c.337C>T (p.Arg113Ter)	PCCB	3	136256588	not provided	
NM_001041.3(SI):c.5110C>T (p.Arg1704Ter)	SI	3	164987225	not provided	
NM_003242.5(TGFBR2):c.1336G>A (p.Asp446Asn)	TGFBR2	3	30674186	not provided	
NM_000249.3(MLH1):c.1997G>A (p.Trp666Ter)	MLH1	3	37048911	not provided	
NM_001904.3(CTNNB1):c.1603C>T (p.Arg535Ter)	CTNNB1	3	41234217	not provided	
NM_002222.5(ITPR1):c.805C>T (p.Arg269Trp)	ITPR1	3	4645678	not provided	
NM_000094.3(COL7A1):c.8329C>T (p.Arg2777Ter)	COL7A1	3	48566539	not provided	
NM_000094.3(COL7A1):c.8012G>A (p.Gly2671Glu)	COL7A1	3	48567608	not provided	
NM_000094.3(COL7A1):c.7865G>A (p.Arg2622Gln)	COL7A1	3	48568100	not provided	

NM_000094.3(COL7A1):c.7723G>A (p.Gly2575Arg)	COL7A1	3	48568819	not provided	
NM_000094.3(COL7A1):c.7442G>A (p.Gly2481Asp)	COL7A1	3	48570177	not provided	
NM_000094.3(COL7A1):c.5300G>A (p.Gly1767Glu)	COL7A1	3	48579376	not provided	
NM_000094.3(COL7A1):c.58C>T (p.Arg20Ter)	COL7A1	3	48595102	not provided	
NM_013334.3(GMPPB):c.260-1G>A	GMPPB	3	49723115	not provided	
NM_001080517.2(SETD5):c.1333C>T (p.Arg445Ter)	SETD5	3	9445193	not provided	
NM_004453.3(ETFDH):c.1832G>A (p.Gly611Glu)	ETFDH	4	158708505	not provided	
NM_001182.4(ALDH7A1):c.834G>A (p.Val278=)	ALDH7A1	5	126568296	not provided	
NM_001182.4(ALDH7A1):c.312+1G>A	ALDH7A1	5	126592663	not provided	
NM_001369.2(DNAH5):c.10441C>T (p.Arg3481Ter)	DNAH5	5	13754317	not provided	
NM_006846.3(SPINK5):c.136C>T (p.Gln46Ter)	SPINK5	5	148070377	not provided	
NM_000806.5(GABRA1):c.640C>T (p.Arg214Cys)	GABRA1	5	161882638	not provided	
NM_002454.2(MTRR):c.1573C>T (p.Arg525Ter)	MTRR	5	7895749	not provided	
NM_032119.3(ADGRV1):c.10213C>T (p.Arg3405Ter)	ADGRV1	5	90728720	not provided	
NM_005654.5(NR2F1):c.262G>A (p.Val88Met)	NR2F1	5	93585285	not provided	
NM_000426.3(LAMA2):c.5476C>T (p.Arg1826Ter)	LAMA2	6	129401254	not provided	
NM_020732.3(ARID1B):c.3025+1G>A	ARID1B	6	157167186	not provided	
NM_020732.3(ARID1B):c.3550+1G>A	ARID1B	6	157184436	not provided	
NM_001069.2(TUBB2A):c.533C>T (p.Thr178Met)	TUBB2A	6	3154668	not provided	
NM_005943.5(MOCS1):c.1102+1G>A	MOCS1	6	39909834	not provided	
NM_000162.3(GCK):c.766G>A (p.Glu256Lys)	GCK	7	44147747	not provided	
NM_000162.3(GCK):c.533G>A (p.Gly178Glu)	GCK	7	44150015	not provided	
NM_001101.3(ACTB):c.617G>A (p.Arg206Gln)	ACTB	7	5528466	not provided	
NM_194456.1(KRIT1):c.1267C>T (p.Arg423Ter)	KRIT1	7	92222966	not provided	
NM_194456.1(KRIT1):c.1146+1G>A	KRIT1	7	92226525	not provided	
NM_194456.1(KRIT1):c.802C>T (p.Gln268Ter)	KRIT1	7	92234851	not provided	
NM_000466.2(PEX1):c.2614C>T (p.Arg872Ter)	PEX1	7	92499808	not provided	
NM_000089.3(COL1A2):c.577G>A (p.Gly193Ser)	COL1A2	7	94406286	not provided	
NM_000089.3(COL1A2):c.1658G>A (p.Gly553Asp)	COL1A2	7	94413940	not provided	
NM_000089.3(COL1A2):c.2567G>A (p.Gly856Glu)	COL1A2	7	94424337	not provided	
NM_000127.2(EXT1):c.1019G>A (p.Arg340His)	EXT1	8	117837145	not provided	
NM_000127.2(EXT1):c.803G>A (p.Gly268Glu)	EXT1	8	118110244	not provided	
NM_000127.2(EXT1):c.535C>T (p.Gln179Ter)	EXT1	8	118110512	not provided	
NM_152419.2(HGSNAT):c.1411G>A (p.Glu471Lys)	HGSNAT	8	43193790	not provided	
NM_017780.3(CHD7):c.151C>T (p.Gln51Ter)	CHD7	8	60741583	not provided	
NM_017780.3(CHD7):c.550C>T (p.Gln184Ter)	CHD7	8	60741982	not provided	
NM_017780.3(CHD7):c.1714C>T (p.Gln572Ter)	CHD7	8	60781048	not provided	
NM_017780.3(CHD7):c.6613C>T (p.Gln2205Ter)	CHD7	8	60853338	not provided	
NM_000318.2(PEX2):c.740G>A (p.Cys247Tyr)	PEX2	8	76983439	not provided	
NM_017890.4(VPS13B):c.1915C>T (p.Arg639Ter)	VPS13B	8	99147912	not provided	
NM_002316.3(LMX1B):c.227G>A (p.Trp76Ter)	LMX1B	9	126615470	not provided	
NM_002316.3(LMX1B):c.781C>T (p.Arg261Cys)	LMX1B	9	126693563	not provided	
NM_000118.3(ENG):c.277C>T (p.Arg93Ter)	ENG	9	127829770	not provided	
NM_000368.4(TSC1):c.2593C>T (p.Gln865Ter)	TSC1	9	132900747	not provided	
NM_020822.2(KCNT1):c.1420C>T (p.Arg474Cys)	KCNT1	9	135768847	not provided	

NM_024757.4(EHMT1):c.2867+5G>A	EHMT1	9	137811620	not provided	
NM_003383.4(VLDLR):c.1586G>A (p.Trp529Ter)	VLDLR	9	2646435	not provided	
NM_000197.1(HSD17B3):c.845C>T (p.Pro282Leu)	HSD17B3	9	96235548	not provided	
NM_147191.1(MMP21):c.1372C>T (p.Arg458Ter)	MMP21	10	125767570	not provided	
NM_001081.3(CUBN):c.2594G>A (p.Ser865Asn)	CUBN	10	17071457	not provided	
NM_006077.3(MICU1):c.553C>T (p.Gln185Ter)	MICU1	10	72508260	not provided	
NM_000043.5(FAS):c.443+1G>A	FAS	10	89008998	not provided	
NM_005097.3(LGI1):c.931C>T (p.Arg311Ter)	LGI1	10	93797060	not provided	
NM_207122.1(EXT2):c.539G>A (p.Trp180Ter)	EXT2	11	44109196	not provided	
NM_207122.1(EXT2):c.544C>T (p.Arg182Ter)	EXT2	11	44109201	not provided	
NM_207122.1(EXT2):c.743+1G>A	EXT2	11	44114302	not provided	
NM_207122.1(EXT2):c.937C>T (p.Gln313Ter)	EXT2	11	44124982	not provided	
NM_004183.3(BEST1):c.604C>T (p.Arg202Trp)	BEST1	11	61956966	not provided	
NM_004183.3(BEST1):c.636+1G>A	BEST1	11	61956999	not provided	
NM_130799.2(MEN1):c.1198C>T (p.Gln400Ter)	MEN1	11	64805186	not provided	
NM_130799.2(MEN1):c.1192C>T (p.Gln398Ter)	MEN1	11	64805192	not provided	
NM_130799.2(MEN1):c.772C>T (p.Gln258Ter)	MEN1	11	64807563	not provided	
NM_130799.2(MEN1):c.660G>A (p.Trp220Ter)	MEN1	11	64807675	not provided	
NM_130799.2(MEN1):c.421C>T (p.Gln141Ter)	MEN1	11	64809689	not provided	
NM_130799.2(MEN1):c.292C>T (p.Arg98Ter)	MEN1	11	64809818	not provided	
NM_001360.2(DHCR7):c.1396G>A (p.Val466Met)	DHCR7	11	71435407	not provided	
NM_000545.6(HNF1A):c.779C>T (p.Thr260Met)	HNF1A	12	120994229	not provided	
NM_000545.6(HNF1A):c.811C>T (p.Arg271Trp)	HNF1A	12	120994261	not provided	
NM_004572.3(PKP2):c.1511-1G>A	PKP2	12	32841206	not provided	
NM_001844.4(COL2A1):c.2833G>A (p.Gly945Ser)	COL2A1	12	47978659	not provided	
NM_006009.3(TUBA1A):c.367C>T (p.Arg123Cys)	TUBA1A	12	49186318	not provided	
NM_000424.3(KRT5):c.1675C>T (p.Arg559Ter)	KRT5	12	52515040	not provided	
NM_000424.3(KRT5):c.555+1G>A	KRT5	12	52519741	not provided	
NM_015665.5(AAAS):c.1331+1G>A	AAAS	12	53308051	not provided	
NM_015665.5(AAAS):c.1159C>T (p.Gln387Ter)	AAAS	12	53308457	not provided	
NM_004004.5(GJB2):c.506G>A (p.Cys169Tyr)	GJB2	13	20189076	not provided	
NM_000059.3(BRCA2):c.1185G>A (p.Trp395Ter)	BRCA2	13	32332663	not provided	
NM_000359.2(TGM1):c.132G>A (p.Trp44Ter)	TGM1	14	24262221	not provided	
NM_019066.4(MAGEL2):c.1996C>T (p.Gln666Ter)	MAGEL2	15	23645747	not provided	
NM_002225.3(IVD):c.1183C>T (p.Arg395Cys)	IVD	15	40418165	not provided	
NM_000138.4(FBN1):c.2581C>T (p.Arg861Ter)	FBN1	15	48495219	not provided	
NM_000138.4(FBN1):c.1042C>T (p.Gln348Ter)	FBN1	15	48520764	not provided	
NM_014985.3(CEP152):c.2038C>T (p.Gln680Ter)	CEP152	15	48767444	not provided	
NM_017882.2(CLN6):c.198+1G>A	CLN6	15	68218535	not provided	
NM_000126.3(ETFA):c.625C>T (p.Arg209Ter)	ETFA	15	76285676	not provided	
NM_001171.5(ABCC6):c.1553G>A (p.Arg518Gln)	ABCC6	16	16190246	not provided	
NM_001083614.1(EARS2):c.1547G>A (p.Arg516Gln)	EARS2	16	23524396	not provided	
NM_004380.2(CREBBP):c.6010C>T (p.Arg2004Ter)	CREBBP	16	3729037	not provided	
NM_004380.2(CREBBP):c.5602C>T (p.Arg1868Trp)	CREBBP	16	3729445	not provided	
NM_016373.3(WWOX):c.410-1G>A	WWOX	16	78164182	not provided	

NM_004483.4(GCSH):c.226C>T (p.Gln76Ter)	GCSH	16	81090603	not provided	
NM_000303.2(PMM2):c.368G>A (p.Arg123Gln)	PMM2	16	8811099	not provided	
NM_000512.4(GALNS):c.860C>T (p.Ser287Leu)	GALNS	16	88835251	not provided	
NM_000512.4(GALNS):c.415G>A (p.Gly139Ser)	GALNS	16	88840999	not provided	
NM_013275.5(ANKRD11):c.1652G>A (p.Trp551Ter)	ANKRD11	16	89284890	not provided	
NM_004278.3(PIGL):c.60G>A (p.Trp20Ter)	PIGL	17	16217286	not provided	
NM_000382.2(ALDH3A2):c.733G>A (p.Asp245Asn)	ALDH3A2	17	19657797	not provided	
NM_000267.3(NF1):c.204+1G>A	NF1	17	31156127	not provided	
NM_000088.3(COL1A1):c.3479G>A (p.Gly1160Asp)	COL1A1	17	50187067	not provided	
NM_000088.3(COL1A1):c.1405C>T (p.Arg469Ter)	COL1A1	17	50194777	not provided	
NM_000088.3(COL1A1):c.757C>T (p.Arg253Ter)	COL1A1	17	50197057	not provided	
NM_000271.4(NPC1):c.1819C>T (p.Arg607Ter)	NPC1	18	23545088	not provided	
NM_000227.4(LAMA3):c.3609+1G>A	LAMA3	18	23928766	not provided	
NM_017653.3(DYM):c.1504C>T (p.Gln502Ter)	DYM	18	49163744	not provided	
NM_001127221.1(CACNA1A):c.1363C>T (p.Arg455Ter)	CACNA1A	19	13317307	not provided	
NM_000215.3(JAK3):c.1765G>A (p.Gly589Ser)	JAK3	19	17837150	not provided	
NM_001972.3(ELANE):c.628G>A (p.Gly210Arg)	ELANE	19	855988	not provided	
NM_000214.2(JAG1):c.3048+1G>A	JAG1	20	10641112	not provided	
NM_000214.2(JAG1):c.148C>T (p.Gln50Ter)	JAG1	20	10672940	not provided	
NM_000022.3(ADA):c.478+1G>A	ADA	20	44625568	not provided	
NM_004975.3(KCNB1):c.934C>T (p.Arg312Cys)	KCNB1	20	49374626	not provided	
NM_000516.5(GNAS):c.784C>T (p.Gln262Ter)	GNAS	20	58909749	not provided	
NM_001958.3(EEF1A2):c.1267C>T (p.Arg423Cys)	EEF1A2	20	63488423	not provided	
NM_000219.5(KCNE1):c.262C>T (p.Gln88Ter)	KCNE1	21	34449373	not provided	
NM_006031.5(PCNT):c.1843C>T (p.Gln615Ter)	PCNT	21	46355533	not provided	
NM_006031.5(PCNT):c.7819C>T (p.Arg2607Ter)	PCNT	21	46430138	not provided	
NM_003073.4(SMARCB1):c.1096C>T (p.Arg366Cys)	SMARCB1	22	23833681	not provided	
NM_006941.3(SOX10):c.425G>A (p.Trp142Ter)	SOX10	22	37983360	not provided	
NM_003560.3(PLA2G6):c.2221C>T (p.Arg741Trp)	PLA2G6	22	38112559	not provided	
NM_003560.3(PLA2G6):c.208C>T (p.Arg70Ter)	PLA2G6	22	38169219	not provided	
NM_000061.2(BTK):c.307C>T (p.Gln103Ter)	BTK	X	101371635	not provided	
NM_000533.4(PLP1):c.191+1G>A	PLP1	X	103785769	not provided	
NM_000533.4(PLP1):c.607G>A (p.Asp203Asn)	PLP1	X	103787951	not provided	
NM_000276.3(OCRL):c.1576C>T (p.Pro526Ser)	OCRL	X	129569373	not provided	
NM_004484.3(GPC3):c.175+1G>A	GPC3	X	133985274	not provided	
NM_000425.4(L1CAM):c.2547+1G>A	L1CAM	X	153865703	not provided	
NM_000425.4(L1CAM):c.2092G>A (p.Gly698Arg)	L1CAM	X	153867401	not provided	
NM_000425.4(L1CAM):c.1453C>T (p.Arg485Ter)	L1CAM	X	153868654	not provided	
NM_000425.4(L1CAM):c.1417C>T (p.Arg473Cys)	L1CAM	X	153868690	not provided	
NM_000425.4(L1CAM):c.523+12C>T	L1CAM	X	153871045	not provided	
NM_000444.5(PHEX):c.1079+1G>A	PHEX	X	22099152	not provided	
NM_000444.5(PHEX):c.1204C>T (p.Gln402Ter)	PHEX	X	22114488	not provided	
NM_000444.5(PHEX):c.1209G>A (p.Trp403Ter)	PHEX	X	22114493	not provided	
NM_004006.2(DMD):c.9148C>T (p.Gln3050Ter)	DMD	X	31348571	not provided	
NM_004006.2(DMD):c.3259C>T (p.Gln1087Ter)	DMD	X	32464603	not provided	

NM_000397.3(CYBB):c.469C>T (p.Arg157Ter)	CYBB	X	37793796	not provided	
NM_000397.3(CYBB):c.868C>T (p.Arg290Ter)	CYBB	X	37801319	not provided	
NM_000397.3(CYBB):c.907C>T (p.His303Tyr)	CYBB	X	37803886	not provided	
NM_000397.3(CYBB):c.1449G>A (p.Trp483Ter)	CYBB	X	37806521	not provided	
NM_003688.3(CASK):c.2100G>A (p.Trp700Ter)	CASK	X	41542746	not provided	
NM_003688.3(CASK):c.609G>A (p.Trp203Ter)	CASK	X	41665376	not provided	
NM_006579.2(EBP):c.439C>T (p.Arg147Cys)	EBP	X	48527255	not provided	
NM_000377.2(WAS):c.223G>A (p.Val75Met)	WAS	X	48684373	not provided	
NM_007075.3(WDR45):c.830+1G>A	WDR45	X	49075363	not provided	
NM_007075.3(WDR45):c.46C>T (p.Gln16Ter)	WDR45	X	49078050	not provided	
NM_005183.3(CACNA1F):c.1873C>T (p.Arg625Ter)	CACNA1F	X	49224798	not provided	
NM_005183.3(CACNA1F):c.148C>T (p.Arg50Ter)	CACNA1F	X	49231805	not provided	
NM_004187.3(KDM5C):c.3880C>T (p.Gln1294Ter)	KDM5C	X	53194297	not provided	
NM_000044.4(AR):c.2257C>T (p.Arg753Ter)	AR	X	67717561	not provided	
NM_001399.4(EDA):c.706+1G>A	EDA	X	70028037	not provided	
NM_001399.4(EDA):c.707-1G>A	EDA	X	70029503	not provided	
NM_001399.4(EDA):c.1069C>T (p.Arg357Trp)	EDA	X	70035502	not provided	
NM_000216.3(ANOS1):c.1891C>T (p.Arg631Ter)	ANOS1	X	8534412	not provided	
NM_002016.1(FLG):c.6109C>T (p.Arg2037Ter)	FLG	1	152308777	not provided	
NM_005518.3(HMGCS2):c.862C>T (p.Arg288Ter)	HMGCS2	1	119757427	not provided	
NM_002016.1(FLG):c.4786C>T (p.Gln1596Ter)	FLG	1	152310100	not provided	
NM_002016.1(FLG):c.1063C>T (p.Gln355Ter)	FLG	1	152313823	not provided	
NM_024408.3(NOTCH2):c.6787C>T (p.Gln2263Ter)	NOTCH2	1	119915935	not provided	
NM_002016.1(FLG):c.11452C>T (p.Gln3818Ter)	FLG	1	152303434	not provided	
NM_004431.4(EPHA2):c.2826-9G>A	EPHA2	1	16125329	not provided	
NM_015100.3(POGZ):c.2350C>T (p.Arg784Ter)	POGZ	1	151408125	not provided	
NM_020699.3(GATAD2B):c.3466C>T (p.Arg1116Ter)	GATAD2B	1	153819725	not provided	
NM_002016.1(FLG):c.2427G>A (p.Trp809Ter)	FLG	1	152312459	not provided	
NM_002016.1(FLG):c.2218C>T (p.Arg740Ter)	FLG	1	152312668	not provided	
NM_015284.3(SZT2):c.4225C>T (p.Arg1409Ter)	SZT2	1	43430098	not provided	
NM_024529.4(CDC73):c.226C>T (p.Arg76Ter)	CDC73	1	193125206	not provided	
NM_024529.4(CDC73):c.355C>T (p.Gln119Ter)	CDC73	1	193135438	not provided	
NM_172362.2(KCNH1):c.1070G>A (p.Arg357Gln)	KCNH1	1	210920032	not provided	
NM_000081.3(LYST):c.10345C>T (p.Arg3449Ter)	LYST	1	235702776	not provided	
NM_005516.2(SLC2A1):c.1199G>A (p.Arg400His)	SLC2A1	1	42927684	not provided	
NM_016343.3(CENPF):c.7219C>T (p.Arg2407Ter)	CENPF	1	214646789	not provided	
NM_000350.2(ABCA4):c.853C>T (p.Gln285Ter)	ABCA4	1	94083357	not provided	
NM_145861.2(EDARADD):c.196C>T (p.Arg66Ter)	EDARADD	1	236427427	not provided	
NM_014795.3(ZEB2):c.3046C>T (p.Arg1016Ter)	ZEB2	2	144396433	not provided	
NM_031844.2(HNRNPU):c.520C>T (p.Gln174Ter)	HNRNPU	1	244863788	not provided	
NM_020451.2(SELENON):c.872G>A (p.Arg291Gln)	SELENON	1	25809150	not provided	
NM_001029882.3(AHDC1):c.1759C>T (p.Arg587Ter)	AHDC1	1	27550357	not provided	
NM_020451.2(SELENON):c.1090C>T (p.Gln364Ter)	SELENON	1	25811533	not provided	
NM_006015.4(ARID1A):c.2988+1G>A	ARID1A	1	26766567	not provided	
NM_015215.3(CAMTA1):c.4231C>T (p.Arg1411Ter)	CAMTA1	1	7744883	not provided	

NM_001029882.3(AHDC1):c.2908C>T (p.Gln970Ter)	AHDC1	1	27549208	not provided	
NM_001271208.1(NEB):c.11806-1G>A	NEB	2	151610867	not provided	
NM_005378.5(MYCN):c.73C>T (p.Gln25Ter)	MYCN	2	15942137	not provided	
NM_021007.2(SCN2A):c.4777G>A (p.Gly1593Arg)	SCN2A	2	165386971	not provided	
NM_022356.3(P3H1):c.1569+1G>A	P3H1	1	42752273	not provided	
NM_001165963.1(SCN1A):c.2797C>T (p.His933Tyr)	SCN1A	2	166037925	not provided	
NM_001204.6(BMPR2):c.637C>T (p.Arg213Ter)	BMPR2	2	202518837	not provided	
NM_001271208.1(NEB):c.24219+1G>A	NEB	2	151499297	not provided	
NM_015265.3(SATB2):c.1285C>T (p.Arg429Ter)	SATB2	2	199328799	not provided	
NM_015025.3(MYT1L):c.1699C>T (p.Arg567Ter)	MYT1L	2	1912024	not provided	
NM_201548.4(CERK1):c.1012C>T (p.Arg338Ter)	CERKL	2	181548741	not provided	
NM_005214.4(CTLA4):c.60G>A (p.Trp20Ter)	CTLA4	2	203868002	not provided	
NM_015265.3(SATB2):c.1286G>A (p.Arg429Gln)	SATB2	2	199328798	not provided	
NM_181457.3(PAX3):c.784C>T (p.Arg262Ter)	PAX3	2	222232086	not provided	
NM_004321.7(KIF1A):c.761G>A (p.Arg254Gln)	KIF1A	2	240783776	not provided	
NM_181457.3(PAX3):c.812G>A (p.Arg271His)	PAX3	2	222221368	not provided	
NM_003042.3(SLC6A1):c.1648G>A (p.Gly550Arg)	SLC6A1	3	11034651	not provided	
NM_000388.3(CASR):c.164C>T (p.Pro55Leu)	CASR	3	122254353	not provided	
NM_006218.3(PIK3CA):c.323G>A (p.Arg108His)	PIK3CA	3	179199148	not provided	
NM_003106.3(SOX2):c.181C>T (p.Gln61Ter)	SOX2	3	181712541	not provided	
NM_198056.2(SCN5A):c.467G>A (p.Trp156Ter)	SCN5A	3	38622415	not provided	
NM_000094.3(COL7A1):c.6769G>A (p.Gly2257Arg)	COL7A1	3	48572924	not provided	
NM_000094.3(COL7A1):c.3140-1G>A	COL7A1	3	48587109	not provided	
NM_000094.3(COL7A1):c.1573C>T (p.Arg525Ter)	COL7A1	3	48591527	not provided	
NM_000388.3(CASR):c.1630C>T (p.Arg544Ter)	CASR	3	122282134	not provided	
NM_005677.3(COLQ):c.679C>T (p.Arg227Ter)	COLQ	3	15470574	not provided	
NM_139248.2(LIPH):c.328C>T (p.Arg110Ter)	LIPH	3	185534854	not provided	
NM_000094.3(COL7A1):c.6190G>A (p.Gly2064Arg)	COL7A1	3	48575233	not provided	
NM_000094.3(COL7A1):c.553C>T (p.Arg185Ter)	COL7A1	3	48593231	not provided	
NM_001904.3(CTNNB1):c.198G>A (p.Trp66Ter)	CTNNB1	3	41224710	not provided	
NM_003722.4(TP63):c.1027C>T (p.Arg343Trp)	TP63	3	189868614	not provided	
NM_133330.2(NSD2):c.2935C>T (p.Arg979Ter)	NSD2	4	1957986	not provided	
NM_015175.2(NBEAL2):c.3592C>T (p.Gln1198Ter)	NBEAL2	3	46999363	not provided	
NM_025132.3(WDR19):c.2363+1G>A	WDR19	4	39234876	not provided	
NM_022455.4(NSD1):c.1288C>T (p.Gln430Ter)	NSD1	5	177209687	not provided	
NM_000094.3(COL7A1):c.5027G>A (p.Gly1676Glu)	COL7A1	3	48580606	not provided	
NM_000094.3(COL7A1):c.4448G>A (p.Gly1483Asp)	COL7A1	3	48583161	not provided	
NM_022455.4(NSD1):c.5842C>T (p.Arg1948Cys)	NSD1	5	177280784	not provided	
NM_022455.4(NSD1):c.5854C>T (p.Arg1952Trp)	NSD1	5	177280796	not provided	
NM_000128.3(F11):c.1489C>T (p.Arg497Ter)	F11	4	186286423	not provided	
NM_003023.4(SH3BP2):c.1252C>T (p.Pro418Ser)	SH3BP2	4	2831581	not provided	
NM_147127.4(EVC2):c.2476C>T (p.Arg826Ter)	EVC2	4	5622562	not provided	
NM_020732.3(ARID1B):c.5338C>T (p.Gln1780Ter)	ARID1B	6	157206479	not provided	
NM_000297.3(PKD2):c.1094+1G>A	PKD2	4	88038502	not provided	
NM_001332.3(CTNND2):c.2653C>T (p.Arg885Ter)	CTNND2	5	11082831	not provided	

NM_022464.4(SIL1):c.1030-9G>A	SIL1	5	138947482	not provided
NM_001182.4(ALDH7A1):c.1489+5G>A	ALDH7A1	5	126549924	not provided
NM_004722.3(AP4M1):c.1321C>T (p.Arg441Ter)	AP4M1	7	100106841	not provided
NM_003060.3(SLC22A5):c.395G>A (p.Trp132Ter)	SLC22A5	5	132378379	not provided
NM_031443.3(CCM2):c.214C>T (p.Gln72Ter)	CCM2	7	45063927	not provided
NM_015570.3(AUTS2):c.1974G>A (p.Trp658Ter)	AUTS2	7	70777144	not provided
NM_001135243.1(TCOF1):c.1278+1G>A	TCOF1	5	150374812	not provided
NM_005654.5(NR2F1):c.424C>T (p.Arg142Cys)	NR2F1	5	93585447	not provided
NM_203290.3(POLR1C):c.70-1G>A	POLR1C	6	43517305	not provided
NM_023073.3(C5orf42):c.57G>A (p.Trp19Ter)	C5orf42	5	37247642	not provided
NM_017780.3(CHD7):c.4189C>T (p.Gln1397Ter)	CHD7	8	60837671	not provided
NM_020732.3(ARID1B):c.5811G>A (p.Trp1937Ter)	ARID1B	6	157206952	not provided
NM_001453.2(FOXC1):c.456G>A (p.Trp152Ter)	FOXC1	6	1610901	not provided
NM_183227.2(RAB23):c.82C>T (p.Arg28Ter)	RAB23	6	57210299	not provided
NM_001080508.2(TBX18):c.1285C>T (p.Arg429Ter)	TBX18	6	84737224	not provided
NM_017890.4(VPS13B):c.8254C>T (p.Gln2752Ter)	VPS13B	8	99817621	not provided
NM_018718.2(CEP41):c.418C>T (p.Gln140Ter)	CEP41	7	130404568	not provided
NM_020632.2(ATP6V0A4):c.334C>T (p.Gln112Ter)	ATP6V0A4	7	138762983	not provided
NM_000083.2(CLCN1):c.854G>A (p.Gly285Glu)	CLCN1	7	143330772	not provided
NM_000093.4(COL5A1):c.3397C>T (p.Arg1133Ter)	COL5A1	9	134809213	not provided
NM_024757.4(EHMT1):c.2704C>T (p.Arg902Ter)	EHMT1	9	137800976	not provided
NM_203447.3(DOCK8):c.949C>T (p.Arg317Ter)	DOCK8	9	328076	not provided
NM_007126.3(VCP):c.475C>T (p.Arg159Cys)	VCP	9	35065352	not provided
NM_016628.4(WAC):c.451C>T (p.Arg151Ter)	WAC	10	28589805	not provided
NM_003919.2(SGCE):c.299G>A (p.Trp100Ter)	SGCE	7	94628293	not provided
NM_014141.5(CNTNAP2):c.1819C>T (p.Gln607Ter)	CNTNAP2	7	147562179	not provided
NM_012330.3(KAT6B):c.3147G>A (p.Pro1049=)	KAT6B	10	75022006	not provided
NM_023110.2(FGFR1):c.1431-1G>A	FGFR1	8	38417992	not provided
NM_000089.3(COL1A2):c.2054G>A (p.Gly685Asp)	COL1A2	7	94419526	not provided
NM_006766.4(KAT6A):c.3553C>T (p.Gln1185Ter)	KAT6A	8	41934667	not provided
NM_017780.3(CHD7):c.434G>A (p.Trp145Ter)	CHD7	8	60741866	not provided
NM_000037.3(ANK1):c.2736-1G>A	ANK1	8	41696588	not provided
NM_012330.3(KAT6B):c.5101C>T (p.Gln1701Ter)	KAT6B	10	75029925	not provided
NM_004820.4(CYP7B1):c.793C>T (p.Gln265Ter)	CYP7B1	8	64615748	not provided
NM_033100.3(CDHR1):c.863-1G>A	CDHR1	10	84205826	not provided
NM_000118.3(ENG):c.816G>A (p.Trp272Ter)	ENG	9	127825231	not provided
NM_000019.3(ACAT1):c.890C>T (p.Thr297Met)	ACAT1	11	108142500	not provided
NM_078480.2(PUF60):c.485G>A (p.Trp162Ter)	PUF60	8	143818398	not provided
NM_015046.5(SETX):c.4936C>T (p.Gln1646Ter)	SETX	9	132326662	not provided
NM_017780.3(CHD7):c.5029C>T (p.Arg1677Ter)	CHD7	8	60845042	not provided
NM_015046.5(SETX):c.4816C>T (p.Arg1606Ter)	SETX	9	132326782	not provided
NM_000503.5(EYA1):c.418+1G>A	EYA1	8	71321733	not provided
NM_017780.3(CHD7):c.934C>T (p.Arg312Ter)	CHD7	8	60742366	not provided
NM_017780.3(CHD7):c.3655C>T (p.Arg1219Ter)	CHD7	8	60830454	not provided
NM_203447.3(DOCK8):c.2606-1G>A	DOCK8	9	382512	not provided

NM_000280.4(PAX6):c.781C>T (p.Arg261Ter)	PAX6	11	31793787	not provided	
NM_000429.2(MAT1A):c.776C>T (p.Ala259Val)	MAT1A	10	80275192	not provided	
NM_004055.4(CAPN5):c.865C>T (p.Arg289Trp)	CAPN5	11	77115560	not provided	
NM_015335.4(MED13L):c.5278C>T (p.Arg1760Ter)	MED13L	12	115980836	not provided	
NM_001130103.1(COL13A1):c.271C>T (p.Arg91Ter)	COL13A1	10	69802694	not provided	
NM_001844.4(COL2A1):c.2536G>A (p.Gly846Arg)	COL2A1	12	47980643	not provided	
NM_004408.3(DNM1):c.709C>T (p.Arg237Trp)	DNM1	9	128220201	not provided	
NM_003172.3(SURF1):c.688C>T (p.Arg230Ter)	SURF1	9	133352509	not provided	
NM_000314.6(PTEN):c.634+5G>A	PTEN	10	87952264	not provided	
NM_130466.3(UBE3B):c.941-1G>A	UBE3B	12	109499632	not provided	
NM_004523.3(KIF11):c.1702+1G>A	KIF11	10	92632694	not provided	
NM_001845.5(COL4A1):c.2672G>A (p.Gly891Asp)	COL4A1	13	110177886	not provided	
NM_000017.3(ACADS):c.820G>A (p.Gly274Ser)	ACADS	12	120738557	not provided	
NM_002336.2(LRP6):c.2953C>T (p.Arg985Ter)	LRP6	12	12150877	not provided	
NM_001844.4(COL2A1):c.1996-1G>A	COL2A1	12	47983439	not provided	
NM_003070.4(SMARCA2):c.3439G>A (p.Asp1147Asn)	SMARCA2	9	2110400	not provided	
NM_001844.4(COL2A1):c.544C>T (p.Gln182Ter)	COL2A1	12	47996613	not provided	
NM_001170629.1(CHD8):c.3733C>T (p.Arg1245Ter)	CHD8	14	21402485	not provided	
NM_007126.3(VCP):c.283C>T (p.Arg95Cys)	VCP	9	35067910	not provided	
NM_020529.2(NFKBIA):c.25C>T (p.Gln9Ter)	NFKBIA	14	35404620	not provided	
NM_025114.3(CEP290):c.1936C>T (p.Gln646Ter)	CEP290	12	88114536	not provided	
NM_000256.3(MYBPC3):c.906-1G>A	MYBPC3	11	47347030	not provided	
NM_001360.2(DHCR7):c.740C>T (p.Ala247Val)	DHCR7	11	71438970	not provided	
NM_024079.4(ALG8):c.1090C>T (p.Arg364Ter)	ALG8	11	78106895	not provided	
NM_018451.4(CENPJ):c.40C>T (p.Gln14Ter)	CENPJ	13	24912986	not provided	
NM_024570.3(RNASEH2B):c.3G>A (p.Met1Ile)	RNASEH2B	13	50910079	not provided	
NM_015335.4(MED13L):c.1336C>T (p.Gln446Ter)	MED13L	12	116009077	not provided	
NM_002693.2(POLG):c.1433+1G>A	POLG	15	89327166	not provided	
NM_002693.2(POLG):c.202C>T (p.Gln68Ter)	POLG	15	89333553	not provided	
NM_001271.3(CHD2):c.1481G>A (p.Trp494Ter)	CHD2	15	92949055	not provided	
NM_001197104.1(KMT2A):c.3991C>T (p.Gln1331Ter)	KMT2A	11	118482071	not provided	
NM_000161.2(GCH1):c.159G>A (p.Trp53Ter)	GCH1	14	54902505	not provided	
NM_000062.2(SERPING1):c.52-1G>A	SERPING1	11	57599878	not provided	
NM_004572.3(PKP2):c.658C>T (p.Gln220Ter)	PKP2	12	32878222	not provided	
NM_001844.4(COL2A1):c.2825G>A (p.Gly942Asp)	COL2A1	12	47978667	not provided	
NM_001844.4(COL2A1):c.2618G>A (p.Gly873Glu)	COL2A1	12	47980561	not provided	
NM_001844.4(COL2A1):c.1546G>A (p.Gly516Ser)	COL2A1	12	47985947	not provided	
NM_002693.2(POLG):c.2419C>T (p.Arg807Cys)	POLG	15	89322749	not provided	
NM_014714.3(IFT140):c.1010-1G>A	IFT140	16	1586276	not provided	
NM_003482.3(KMT2D):c.16501C>T (p.Arg5501Ter)	KMT2D	12	49022063	not provided	
NM_003482.3(KMT2D):c.16184G>A (p.Trp5395Ter)	KMT2D	12	49022744	not provided	
NM_000088.3(COL1A1):c.3415G>A (p.Gly1139Ser)	COL1A1	17	50187492	not provided	
NM_003482.3(KMT2D):c.15143G>A (p.Arg5048His)	KMT2D	12	49026823	not provided	
NM_003482.3(KMT2D):c.13999+1G>A	KMT2D	12	49030279	not provided	
NM_000359.2(TGM1):c.379C>T (p.Arg127Ter)	TGM1	14	24261824	not provided	

NM_003482.3(KMT2D):c.5707C>T (p.Arg1903Ter)	KMT2D	12	49042816	not provided	
NM_002905.3(RDH5):c.160C>T (p.Arg54Ter)	RDH5	12	55721344	not provided	
NM_001079668.2(NKX2-1):c.637C>T (p.Gln213Ter)	NKX2-1	14	36517847	not provided	
NM_024110.4(CARD14):c.349+1G>A	CARD14	17	80182791	not provided	
NM_000282.3(PCCA):c.1284+1G>A	PCCA	13	100302999	not provided	
NM_015295.2(SMCHD1):c.1030C>T (p.Arg344Ter)	SMCHD1	18	2694683	not provided	
NM_001942.3(DSG1):c.724C>T (p.Arg242Ter)	DSG1	18	31333628	not provided	
NM_013275.5(ANKRD11):c.7552C>T (p.Gln2518Ter)	ANKRD11	16	89275110	not provided	
NM_018451.4(CENPJ):c.1696C>T (p.Arg566Ter)	CENPJ	13	24906342	not provided	
NM_002693.2(POLG):c.925C>T (p.Arg309Cys)	POLG	15	89329041	not provided	
NM_001271.3(CHD2):c.3214C>T (p.Arg1072Ter)	CHD2	15	92984477	not provided	
NM_000135.2(FANCA):c.4011-1G>A	FANCA	16	89739290	not provided	
NM_000548.4(TSC2):c.4183C>T (p.Gln1395Ter)	TSC2	16	2084405	not provided	
NM_001958.3(EEF1A2):c.370G>A (p.Glu124Lys)	EEF1A2	20	63495056	not provided	
NM_004782.3(SNAP29):c.85C>T (p.Arg29Ter)	SNAP29	22	20859195	not provided	
NM_020366.3(RPGRIP1):c.2668C>T (p.Arg890Ter)	RPGRIP1	14	21326131	not provided	
NM_004960.3(FUS):c.1574C>T (p.Pro525Leu)	FUS	16	31191431	not provided	
NM_020461.3(TUBGCP6):c.1833+1G>A	TUBGCP6	22	50226049	not provided	
NM_004380.2(CREBBP):c.6241C>T (p.Gln2081Ter)	CREBBP	16	3728806	not provided	
NM_004380.2(CREBBP):c.3609+1G>A	CREBBP	16	3757808	not provided	
NM_001170629.1(CHD8):c.2992C>T (p.Gln998Ter)	CHD8	14	21405780	not provided	
NM_000061.2(BTK):c.469C>T (p.Gln157Ter)	BTK	X	101362612	not provided	
NM_000359.2(TGM1):c.377G>A (p.Arg126His)	TGM1	14	24261826	not provided	
NM_002734.4(PRKAR1A):c.46C>T (p.Arg16Ter)	PRKAR1A	17	68515445	not provided	
NM_000194.2(HPRT1):c.145C>T (p.Leu49Phe)	HPRT1	X	134475191	not provided	
NM_020988.2(GNAO1):c.119G>A (p.Gly40Glu)	GNAO1	16	56192574	not provided	
NM_001258221.1(KCTD1):c.97C>T (p.His33Tyr)	KCTD1	18	26501139	not provided	
NM_021728.3(OTX2):c.226C>T (p.Arg76Ter)	OTX2	14	56804235	not provided	
NM_004614.4(TK2):c.103C>T (p.Gln35Ter)	TK2	16	66549959	not provided	
NM_001083962.1(TCF4):c.655G>A (p.Asp219Asn)	TCF4	18	55279551	not provided	
NM_013275.5(ANKRD11):c.3562C>T (p.Arg1188Ter)	ANKRD11	16	89282980	not provided	
NM_000455.4(STK11):c.717G>A (p.Trp239Ter)	STK11	19	1220700	not provided	
NM_001271043.2(NFIX):c.690G>A (p.Trp230Ter)	NFIX	19	13073465	not provided	
NM_003119.3(SPG7):c.679C>T (p.Arg227Ter)	SPG7	16	89526389	not provided	
NM_000267.3(NF1):c.2446C>T (p.Arg816Ter)	NF1	17	31229061	not provided	
NM_000444.5(PHEX):c.1543C>T (p.Gln515Ter)	PHEX	X	22178333	not provided	
NM_004247.3(EFTUD2):c.1669C>T (p.Gln557Ter)	EFTUD2	17	44860482	not provided	
NM_000138.4(FBN1):c.5917+1G>A	FBN1	15	48445375	not provided	
NM_152296.4(ATP1A3):c.2840G>A (p.Gly947Glu)	ATP1A3	19	41967743	not provided	
NM_000088.3(COL1A1):c.2119G>A (p.Gly707Ser)	COL1A1	17	50191796	not provided	
NM_015338.5(ASXL1):c.1282C>T (p.Gln428Ter)	ASXL1	20	32433480	not provided	
NM_015339.4(ADNP):c.790C>T (p.Arg264Ter)	ADNP	20	50893924	not provided	
NM_130810.3(DNAAF4):c.1047+1G>A	DNAAF4	15	55434904	not provided	
NM_000781.2(CYP11A1):c.425+1G>A	CYP11A1	15	74347899	not provided	
NM_000444.5(PHEX):c.2239C>T (p.Arg747Ter)	PHEX	X	22247942	not provided	

NM_144773.3(PROKR2):c.458+1G>A	PROKR2	20	5313911	not provided	
NM_014271.3(IL1RAPL1):c.703+1G>A	IL1RAPL1	X	29399309	not provided	
NM_021628.2(ALOXE3):c.1889C>T (p.Pro630Leu)	ALOXE3	17	8103390	not provided	
NM_000397.3(CYBB):c.1272G>A (p.Trp424Ter)	CYBB	X	37805126	not provided	
NM_005993.4(TBCD):c.1423G>A (p.Ala475Thr)	TBCD	17	82870328	not provided	
NM_001271.3(CHD2):c.4413+1G>A	CHD2	15	93004752	not provided	
NM_001111125.2(IQSEC2):c.2984G>A (p.Arg995Gln)	IQSEC2	X	53241815	not provided	
NM_001083962.1(TCF4):c.1471C>T (p.Gln491Ter)	TCF4	18	55234563	not provided	
NM_001958.3(EEF1A2):c.271G>A (p.Asp91Asn)	EEF1A2	20	63495909	not provided	
NM_001396.3(DYRK1A):c.787C>T (p.Arg263Ter)	DYRK1A	21	37490297	not provided	
NM_001848.2(COL6A1):c.842G>A (p.Gly281Glu)	COL6A1	21	45989121	not provided	
NM_000206.2(IL2RG):c.758-1G>A	IL2RG	X	71108696	not provided	
NM_000489.4(ATRX):c.5957-1G>A	ATRX	X	77593850	not provided	
NM_003560.3(PLA2G6):c.1077G>A (p.Ser359=)	PLA2G6	22	38132831	not provided	
NM_001271043.2(NFIX):c.661C>T (p.Gln221Ter)	NFIX	19	13073436	not provided	
NM_001429.3(EP300):c.3472C>T (p.Gln1158Ter)	EP300	22	41157379	not provided	
NM_000548.4(TSC2):c.4594C>T (p.Gln1532Ter)	TSC2	16	2085254	not provided	
NM_000061.2(BTK):c.1573C>T (p.Arg525Ter)	BTK	X	101354688	not provided	
NM_194277.2(FRMD7):c.905+1G>A	FRMD7	X	132082362	not provided	
NM_004750.4(CRLF1):c.322C>T (p.Gln108Ter)	CRLF1	19	18599640	not provided	
NM_018196.3(TMLHE):c.5G>A (p.Trp2Ter)	TMLHE	X	155545272	not provided	
NM_001849.3(COL6A2):c.2422+1G>A	COL6A2	21	46126238	not provided	
NM_000444.5(PHEX):c.733-1G>A	PHEX	X	22093982	not provided	
NM_016327.2(UPB1):c.873+1G>A	UPB1	22	24520469	not provided	
NM_013275.5(ANKRD11):c.7234C>T (p.Gln2412Ter)	ANKRD11	16	89279308	not provided	
NM_000496.2(CRYBB2):c.563G>A (p.Arg188His)	CRYBB2	22	25231717	not provided	
NM_013275.5(ANKRD11):c.1801C>T (p.Arg601Ter)	ANKRD11	16	89284741	not provided	
NM_006086.3(TUBB3):c.689C>T (p.Ser230Leu)	TUBB3	16	89935140	not provided	
NM_000444.5(PHEX):c.1080-1G>A	PHEX	X	22111466	not provided	
NM_144997.5(FLCN):c.1504C>T (p.Gln502Ter)	FLCN	17	17215019	not provided	
NM_000444.5(PHEX):c.1158G>A (p.Trp386Ter)	PHEX	X	22111545	not provided	
NM_000444.5(PHEX):c.1601C>T (p.Pro534Leu)	PHEX	X	22190458	not provided	
NM_000444.5(PHEX):c.1645+1G>A	PHEX	X	22190503	not provided	
NM_000444.5(PHEX):c.2147+1G>A	PHEX	X	22245410	not provided	
NM_000430.3(PAFAH1B1):c.337C>T (p.Arg113Ter)	PAFAH1B1	17	2667136	not provided	
NM_000276.3(OCRL):c.940-11G>A	OCRL	X	129562373	not provided	
NM_001193416.2(DDX3X):c.1804C>T (p.Arg602Ter)	DDX3X	X	41347349	not provided	
NM_018896.4(CACNA1G):c.2881G>A (p.Ala961Thr)	CACNA1G	17	50592063	not provided	
NM_007075.3(WDR45):c.19C>T (p.Arg7Ter)	WDR45	X	49078077	not provided	
NM_020717.3(SHROOM4):c.3739C>T (p.Gln1247Ter)	SHROOM4	X	50607403	not provided	
NM_000152.4(GAA):c.2237G>A (p.Trp746Ter)	GAA	17	80117015	not provided	
NM_001456.3(FLNA):c.4519C>T (p.Gln1507Ter)	FLNA	X	154358524	not provided	
NM_006306.3(SMC1A):c.1192C>T (p.Arg398Ter)	SMC1A	X	53411823	not provided	
NM_004586.2(RPS6KA3):c.1934G>A (p.Trp645Ter)	RPS6KA3	X	20161669	not provided	
NM_000199.3(SGSH):c.629G>A (p.Trp210Ter)	SGSH	17	80214206	not provided	

NM_031407.6(HUWE1):c.9208C>T (p.Arg3070Cys)	HUWE1	X	53551078	not provided	
NM_000044.4(AR):c.1886-1G>A	AR	X	67711401	not provided	
NM_000444.5(PHEX):c.941G>A (p.Trp314Ter)	PHEX	X	22099013	not provided	
NM_001008537.2(KIAA2022):c.1441C>T (p.Arg481Ter)	KIAA2022	X	74743116	not provided	
NM_000390.3(CHM):c.1138C>T (p.Gln380Ter)	CHM	X	85956181	not provided	
NM_000444.5(PHEX):c.1587-1G>A	PHEX	X	22190443	not provided	
NM_152296.4(ATP1A3):c.2324C>T (p.Pro775Leu)	ATP1A3	19	41970482	not provided	
NM_000444.5(PHEX):c.2071-1G>A	PHEX	X	22245332	not provided	
NM_000397.3(CYBB):c.271C>T (p.Arg91Ter)	CYBB	X	37791993	not provided	
NM_015338.5(ASXL1):c.1867C>T (p.Gln623Ter)	ASXL1	20	32434579	not provided	
NM_001039590.2(USP9X):c.1140G>A (p.Trp380Ter)	USP9X	X	41141410	not provided	
NM_003688.3(CASK):c.429+1G>A	CASK	X	41739383	not provided	
NM_000044.4(AR):c.220C>T (p.Gln74Ter)	AR	X	67545366	not provided	
NM_000044.4(AR):c.1847G>A (p.Arg616His)	AR	X	67686088	not provided	
NM_000044.4(AR):c.2566C>T (p.Arg856Cys)	AR	X	67722943	not provided	
NM_001849.3(COL6A2):c.244C>T (p.Gln82Ter)	COL6A2	21	46112107	not provided	
NM_000044.4(AR):c.2668G>A (p.Val890Met)	AR	X	67723746	not provided	
NM_000206.2(IL2RG):c.924+1G>A	IL2RG	X	71108276	not provided	
NM_018486.2(HDAC8):c.787C>T (p.Gln263Ter)	HDAC8	X	72464682	not provided	
NM_000390.3(CHM):c.799C>T (p.Arg267Ter)	CHM	X	85958881	not provided	
NM_000194.2(HPRT1):c.202C>T (p.Leu68Phe)	HPRT1	X	134475248	not provided	
NM_000444.5(PHEX):c.664-1G>A	PHEX	X	22090428	not provided	
NM_000444.5(PHEX):c.1367G>A (p.Trp456Ter)	PHEX	X	22133587	not provided	
NM_000444.5(PHEX):c.1645C>T (p.Arg549Ter)	PHEX	X	22190502	not provided	
NM_000444.5(PHEX):c.1806G>A (p.Trp602Ter)	PHEX	X	22221650	not provided	
NM_000444.5(PHEX):c.1979G>A (p.Trp660Ter)	PHEX	X	22227520	not provided	
NM_000444.5(PHEX):c.2104C>T (p.Arg702Ter)	PHEX	X	22245366	not provided	
NM_000047.2(ARSE):c.1711C>T (p.Gln571Ter)	ARSE	X	2934891	not provided	
NM_000047.2(ARSE):c.1300G>A (p.Gly434Ser)	ARSE	X	2936853	not provided	
NM_001111125.2(IQSEC2):c.2272C>T (p.Arg758Ter)	IQSEC2	X	53250304	not provided	
NM_000044.4(AR):c.1822C>T (p.Arg608Ter)	AR	X	67686063	not provided	
NM_001399.4(EDA):c.872G>A (p.Gly291Glu)	EDA	X	70033476	not provided	
NM_005120.2(MED12):c.5125C>T (p.Arg1709Ter)	MED12	X	71136380	not provided	
NM_001008537.2(KIAA2022):c.1042C>T (p.Arg348Ter)	KIAA2022	X	74743515	not provided	
NM_000390.3(CHM):c.757C>T (p.Arg253Ter)	CHM	X	85958923	not provided	
NM_000033.3(ABCD1):c.1166G>A (p.Arg389His)	ABCD1	X	153736196	not provided	
NM_000272.3(NPHP1):c.329+1G>A	NPHP1	2	110178422	not provided	
NM_002016.1(FLG):c.5230C>T (p.Gln1744Ter)	FLG	1	152309656	not provided	
NM_000433.3(NCF2):c.257+1G>A	NCF2	1	183586894	not provided	
NM_206933.2(USH2A):c.12691C>T (p.Gln4231Ter)	USH2A	1	215675220	not provided	
NM_206933.2(USH2A):c.486-14G>A	USH2A	1	216418693	not provided	
NM_000350.2(ABCA4):c.1988G>A (p.Trp663Ter)	ABCA4	1	94060709	not provided	
NM_001271208.1(NEB):c.17262G>A (p.Trp5754Ter)	NEB	2	151570249	not provided	
NM_018136.4(ASPM):c.2409G>A (p.Trp803Ter)	ASPM	1	197133360	not provided	
NM_001256850.1(TTN):c.95902C>T (p.Arg31968Ter)	TTN	2	178535790	not provided	

NM_000350.2(ABCA4):c.1834C>T (p.Gln612Ter)	ABCA4	1	94062680	not provided	
NM_000348.3(SRD5A2):c.100G>A (p.Gly34Arg)	SRD5A2	2	31580801	not provided	
NM_006516.2(SLC2A1):c.418G>A (p.Val140Met)	SLC2A1	1	42930724	not provided	
NM_000350.2(ABCA4):c.6816+1G>A	ABCA4	1	93996108	not provided	
NM_000350.2(ABCA4):c.3403C>T (p.Gln1135Ter)	ABCA4	1	94041328	not provided	
NM_014795.3(ZEB2):c.2908C>T (p.Gln970Ter)	ZEB2	2	144396571	not provided	
NM_002977.3(SCN9A):c.2108G>A (p.Trp703Ter)	SCN9A	2	166280559	not provided	
NM_001271208.1(NEB):c.9046C>T (p.Arg3016Ter)	NEB	2	151636283	not provided	
NM_000090.3(COL3A1):c.1471C>T (p.Arg491Ter)	COL3A1	2	188995061	not provided	
NM_024665.5(TBL1XR1):c.1108G>A (p.Asp370Asn)	TBL1XR1	3	177038112	not provided	
NM_015265.3(SATB2):c.1196G>A (p.Arg399His)	SATB2	2	199328888	not provided	
NM_025216.2(WNT10A):c.337C>T (p.Arg113Cys)	WNT10A	2	218882384	not provided	
NM_000404.3(GLB1):c.841C>T (p.His281Tyr)	GLB1	3	33051956	not provided	
NM_000348.3(SRD5A2):c.542C>T (p.Pro181Leu)	SRD5A2	2	31531376	not provided	
NM_000094.3(COL7A1):c.5499C>T (p.Gly1833=)	COL7A1	3	48578354	not provided	
NM_000090.3(COL3A1):c.897+1G>A	COL3A1	2	188991532	not provided	
NM_000094.3(COL7A1):c.1732C>T (p.Arg578Ter)	COL7A1	3	48590721	not provided	
NM_015265.3(SATB2):c.868C>T (p.Gln290Ter)	SATB2	2	199349006	not provided	
NM_000094.3(COL7A1):c.5047C>T (p.Arg1683Ter)	COL7A1	3	48580586	not provided	
NM_000094.3(COL7A1):c.682+1G>A	COL7A1	3	48593101	not provided	
NM_000248.3(MITF):c.763C>T (p.Arg255Ter)	MITF	3	69959325	not provided	
NM_145860.1(PDCD10):c.103C>T (p.Arg35Ter)	PDCD10	3	167704889	not provided	
NM_017875.2(SLC25A38):c.166C>T (p.Gln56Ter)	SLC25A38	3	39389591	not provided	
NM_014159.6(SETD2):c.4774C>T (p.Arg1592Ter)	SETD2	3	47106062	not provided	
NM_000094.3(COL7A1):c.7078G>A (p.Gly2360Arg)	COL7A1	3	48571269	not provided	
NM_001099287.1(NIPAL4):c.688G>A (p.Gly230Arg)	NIPAL4	5	157471733	not provided	
NM_000094.3(COL7A1):c.7012C>T (p.Arg2338Ter)	COL7A1	3	48572138	not provided	
NM_000094.3(COL7A1):c.6781C>T (p.Arg2261Ter)	COL7A1	3	48572912	not provided	
NM_000094.3(COL7A1):c.6262G>A (p.Gly2088Arg)	COL7A1	3	48575081	not provided	
NM_006846.3(SPINK5):c.2240+1G>A	SPINK5	5	148118565	not provided	
NM_000094.3(COL7A1):c.7485+5G>A	COL7A1	3	48570129	not provided	
NM_000094.3(COL7A1):c.7344G>A (p.Val2448=)	COL7A1	3	48570639	not provided	
NM_014845.5(FIG4):c.2212C>T (p.Gln738Ter)	FIG4	6	109791407	not provided	
NM_000094.3(COL7A1):c.5344G>A (p.Gly1782Arg)	COL7A1	3	48579241	not provided	
NM_000094.3(COL7A1):c.4373C>T (p.Pro1458Leu)	COL7A1	3	48583584	not provided	
NM_020732.3(ARID1B):c.1360C>T (p.Gln454Ter)	ARID1B	6	156779289	not provided	
NM_021942.5(TRAPPC11):c.2407C>T (p.Gln803Ter)	TRAPPC11	4	183693937	not provided	
NM_003023.4(SH3BP2):c.1258G>A (p.Gly420Arg)	SH3BP2	4	2831587	not provided	
NM_006005.3(WFS1):c.376G>A (p.Ala126Thr)	WFS1	4	6289047	not provided	
NM_003060.3(SLC22A5):c.1252C>T (p.Gln418Ter)	SLC22A5	5	132390889	not provided	
NM_015570.3(AUTS2):c.1483C>T (p.Arg495Ter)	AUTS2	7	70766128	not provided	
NM_000089.3(COL1A2):c.983G>A (p.Gly328Asp)	COL1A2	7	94409769	not provided	
NM_000445.4(PLEC):c.7612C>T (p.Gln2538Ter)	PLEC	8	143922290	not provided	
NM_181523.2(PIK3R1):c.1425+1G>A	PIK3R1	5	68293835	not provided	
NM_000165.4(GJA1):c.605G>A (p.Arg202His)	GJA1	6	121447452	not provided	

NM_032119.3(ADGRV1):c.11410C>T (p.Arg3804Ter)	ADGRV1	5	90755015	not provided	
NM_003165.3(STXBP1):c.1012C>T (p.Gln338Ter)	STXBP1	9	127672099	not provided	
NM_017780.3(CHD7):c.5315G>A (p.Trp1772Ter)	CHD7	8	60849065	not provided	
NM_033305.2(VPS13A):c.799C>T (p.Arg267Ter)	VPS13A	9	77219998	not provided	
NM_004959.4(NR5A1):c.937C>T (p.Arg313Cys)	NR5A1	9	124493083	not provided	
NM_020732.3(ARID1B):c.2776C>T (p.Gln926Ter)	ARID1B	6	157148848	not provided	
NM_006772.2(SYNGAP1):c.490C>T (p.Arg164Ter)	SYNGAP1	6	33432787	not provided	
NM_006772.2(SYNGAP1):c.1716G>A (p.Trp572Ter)	SYNGAP1	6	33440768	not provided	
NM_016628.4(WAC):c.1072C>T (p.Gln358Ter)	WAC	10	28608338	not provided	
NM_014251.2(SLC25A13):c.955C>T (p.Arg319Ter)	SLC25A13	7	96184990	not provided	
NM_000048.3(ASL):c.578G>A (p.Arg193Gln)	ASL	7	66086797	not provided	
NM_016628.4(WAC):c.1556+1G>A	WAC	10	28614686	not provided	
NM_006077.3(MICU1):c.1078-1G>A	MICU1	10	72408038	not provided	
NM_003815.2(ADAM9):c.196-1G>A	ADAM9	8	39011657	not provided	
NM_000314.6(PTEN):c.1027-1G>A	PTEN	10	87965286	not provided	
NM_000218.2(KCNQ1):c.922-1G>A	KCNQ1	11	2583434	not provided	
NM_000256.3(MYBPC3):c.1458G>A (p.Trp486Ter)	MYBPC3	11	47342744	not provided	
NM_001197104.1(KMT2A):c.3464G>A (p.Cys1155Tyr)	KMT2A	11	118478096	not provided	
NM_003070.4(SMARCA2):c.3484C>T (p.Arg1162Cys)	SMARCA2	9	2115849	not provided	
NM_000280.4(PAX6):c.949C>T (p.Arg317Ter)	PAX6	11	31793521	not provided	
NM_000192.3(TBX5):c.670C>T (p.Gln224Ter)	TBX5	12	114385561	not provided	
NM_033305.2(VPS13A):c.3889C>T (p.Arg1297Ter)	VPS13A	9	77302991	not provided	
NM_003482.3(KMT2D):c.12844C>T (p.Arg4282Ter)	KMT2D	12	49031861	not provided	
NM_000256.3(MYBPC3):c.3617G>A (p.Gly1206Asp)	MYBPC3	11	47332576	not provided	
NM_003482.3(KMT2D):c.8366+1G>A	KMT2D	12	49039221	not provided	
NM_003482.3(KMT2D):c.7411C>T (p.Arg2471Ter)	KMT2D	12	49040359	not provided	
NM_006009.3(TUBA1A):c.641G>A (p.Arg214His)	TUBA1A	12	49185725	not provided	
NM_018451.4(CENPJ):c.469C>T (p.Gln157Ter)	CENPJ	13	24912045	not provided	
NM_000280.4(PAX6):c.622C>T (p.Arg208Trp)	PAX6	11	31794690	not provided	
NM_000256.3(MYBPC3):c.3257G>A (p.Trp1086Ter)	MYBPC3	11	47333267	not provided	
NM_014384.2(ACAD8):c.567+1G>A	ACAD8	11	134259085	not provided	
NM_014384.2(ACAD8):c.958G>A (p.Ala320Thr)	ACAD8	11	134261756	not provided	
NM_001376.4(DYNC1H1):c.2327C>T (p.Pro776Leu)	DYNC1H1	14	101986552	not provided	
NM_000020.2(ACVRL1):c.1435C>T (p.Arg479Ter)	ACVRL1	12	51920816	not provided	
NM_000260.3(MYO7A):c.3724C>T (p.Gln1242Ter)	MYO7A	11	77190113	not provided	
NM_000359.2(TGM1):c.790C>T (p.Arg264Trp)	TGM1	14	24260026	not provided	
NM_130799.2(MEN1):c.913-1G>A	MEN1	11	64806369	not provided	
NM_000545.6(HNF1A):c.526C>T (p.Gln176Ter)	HNF1A	12	120989032	not provided	
NM_002834.4(PTPN11):c.1051C>T (p.Arg351Ter)	PTPN11	12	112477974	not provided	
NM_000719.6(CACNA1C):c.1553G>A (p.Arg518His)	CACNA1C	12	2566466	not provided	
NM_025114.3(CEP290):c.451C>T (p.Arg151Ter)	CEP290	12	88131209	not provided	
NM_002693.2(POLG):c.823C>T (p.Arg275Ter)	POLG	15	89330113	not provided	
NM_001271.3(CHD2):c.340C>T (p.Arg114Ter)	CHD2	15	92927289	not provided	
NM_001271.3(CHD2):c.1462C>T (p.Gln488Ter)	CHD2	15	92949036	not provided	
NM_000359.2(TGM1):c.1417G>A (p.Gly473Ser)	TGM1	14	24256063	not provided	

NM_002225.3(IVD):c.559+1G>A	IVD	15	40411354	not provided	
NM_000359.2(TGM1):c.427C>T (p.Arg143Cys)	TGM1	14	24261776	not provided	
NM_016373.3(WWOX):c.131G>A (p.Trp44Ter)	WWOX	16	78108446	not provided	
NM_016630.6(SPG21):c.736C>T (p.Arg246Ter)	SPG21	15	64965394	not provided	
NM_000267.3(NF1):c.1591C>T (p.Gln531Ter)	NF1	17	31219068	not provided	
NM_001171.5(ABCC6):c.1132C>T (p.Gln378Ter)	ABCC6	16	16202045	not provided	
NM_033419.4(PGAP3):c.694+1G>A	PGAP3	17	39673513	not provided	
NM_152594.2(SPRED1):c.46C>T (p.Arg16Ter)	SPRED1	15	38299386	not provided	
NM_000548.4(TSC2):c.4493+1G>A	TSC2	16	2084716	not provided	
NM_000088.3(COL1A1):c.1354-12G>A	COL1A1	17	50194840	not provided	
NM_002968.2(SALL1):c.1393C>T (p.Gln465Ter)	SALL1	16	51140829	not provided	
NM_000267.3(NF1):c.4773-1G>A	NF1	17	31325819	not provided	
NM_000458.3(HNF1B):c.541C>T (p.Arg181Ter)	HNF1B	17	37739443	not provided	
NM_000088.3(COL1A1):c.3208G>A (p.Gly1070Ser)	COL1A1	17	50188149	not provided	
NM_002474.2(MYH11):c.4578+1G>A	MYH11	16	15721421	not provided	
NM_001139.2(ALOX12B):c.530G>A (p.Trp177Ter)	ALOX12B	17	8080778	not provided	
NM_000346.3(SOX9):c.1180C>T (p.Arg394Ter)	SOX9	17	72124037	not provided	
NM_003244.3(TGIF1):c.269G>A (p.Arg90His)	TGIF1	18	3457390	not provided	
NM_001271043.2(NFIX):c.583+1G>A	NFIX	19	13025553	not provided	
NM_000267.3(NF1):c.669G>A (p.Trp223Ter)	NF1	17	31181724	not provided	
NM_145068.3(TRPV3):c.1703G>A (p.Gly568Asp)	TRPV3	17	3524238	not provided	
NM_003079.4(SMARCE1):c.751C>T (p.Arg251Ter)	SMARCE1	17	40631657	not provided	
NM_152296.4(ATP1A3):c.2116G>A (p.Gly706Arg)	ATP1A3	19	41975776	not provided	
NM_001972.3(ELANE):c.137C>T (p.Ser46Phe)	ELANE	19	852945	not provided	
NM_001396.3(DYRK1A):c.349C>T (p.Arg117Ter)	DYRK1A	21	37480659	not provided	
NM_003073.4(SMARCB1):c.1121G>A (p.Arg374Gln)	SMARCB1	22	23834143	not provided	
NM_000455.4(STK11):c.735-1G>A	STK11	19	1221212	not provided	
NM_000061.2(BTK):c.1103G>A (p.Gly368Glu)	BTK	X	101357583	not provided	
NM_000116.4(TAZ):c.699+1G>A	TAZ	X	154420265	not provided	
NM_000330.3(RS1):c.208G>A (p.Gly70Ser)	RS1	X	18647309	not provided	
NM_000444.5(PHEX):c.1483-1G>A	PHEX	X	22178272	not provided	
NM_003159.2(CDKL5):c.1006C>T (p.Gln336Ter)	CDKL5	X	18603930	not provided	
NM_000330.3(RS1):c.668G>A (p.Cys223Tyr)	RS1	X	18642011	not provided	
NM_000444.5(PHEX):c.500G>A (p.Trp167Ter)	PHEX	X	22077539	not provided	
NM_000444.5(PHEX):c.1180C>T (p.Gln394Ter)	PHEX	X	22114464	not provided	
NM_004006.2(DMD):c.1602+1G>A	DMD	X	32595756	not provided	
NM_001849.3(COL6A2):c.853+1G>A	COL6A2	21	46115926	not provided	
NM_000487.5(ARSA):c.979G>A (p.Gly327Ser)	ARSA	22	50626154	not provided	
NM_002049.3(GATA1):c.622G>A (p.Gly208Arg)	GATA1	X	48792346	not provided	
NM_000444.5(PHEX):c.871C>T (p.Arg291Ter)	PHEX	X	22096976	not provided	
NM_014009.3(FOXP3):c.1009C>T (p.Arg337Ter)	FOXP3	X	49253161	not provided	
NM_003688.3(CASK):c.2368C>T (p.Gln790Ter)	CASK	X	41531144	not provided	
NM_000206.2(IL2RG):c.43C>T (p.Gln15Ter)	IL2RG	X	71111497	not provided	
NM_000390.3(CHM):c.1213C>T (p.Gln405Ter)	CHM	X	85911292	not provided	
NM_004006.2(DMD):c.2168+1G>A	DMD	X	32545158	not provided	

NM_000377.2(WAS):c.559+5G>A	WAS	X	48686139	not provided	
NM_000390.3(CHM):c.1342C>T (p.Gln448Ter)	CHM	X	85901091	not provided	
NM_001846.3(COL4A2):c.3490G>A (p.Gly1164Arg)	COL4A2	13	110492105	not provided	
NM_001197104.1(KMT2A):c.11230C>T (p.Arg3744Ter)	KMT2A	11	118519701	not provided	
NM_000019.3(ACAT1):c.622C>T (p.Arg208Ter)	ACAT1	11	108140107	not provided	
NM_001972.3(ELANE):c.170C>T (p.Ala57Val)	ELANE	19	852978	not provided	
NM_000052.6(ATP7A):c.2867G>A (p.Trp956Ter)	ATP7A	X	78021030	not provided	
NM_023110.2(FGFR1):c.1468G>A (p.Gly490Arg)	FGFR1	8	38417954	not provided	
NM_138694.3(PKHD1):c.5912G>A (p.Gly1971Asp)	PKHD1	6	51934319	not provided	
NM_138694.3(PKHD1):c.2414C>T (p.Pro805Leu)	PKHD1	6	52046182	not provided	
NM_000314.6(PTEN):c.254-1G>A	PTEN	10	87933012	not provided	
NM_002016.1(FLG):c.7249C>T (p.Gln2417Ter)	FLG	1	152307637	not provided	
NM_020699.3(GATAD2B):c.658C>T (p.Gln220Ter)	GATAD2B	1	153818111	not provided	
NM_019032.5(ADAMTSL4):c.421C>T (p.Arg141Ter)	ADAMTSL4	1	150353240	not provided	
NM_020699.3(GATAD2B):c.1045C>T (p.Gln349Ter)	GATAD2B	1	153816444	not provided	
NM_206933.2(USH2A):c.12276>A (p.Trp409Ter)	USH2A	1	216324269	not provided	
NM_018060.3(IARS2):c.2175+1G>A	IARS2	1	220138044	not provided	
NM_006147.3(IRF6):c.454C>T (p.Gln152Ter)	IRF6	1	209795344	not provided	
NM_006147.3(IRF6):c.352C>T (p.Gln118Ter)	IRF6	1	209796375	not provided	
NM_014053.3(FLVCR1):c.1093-1G>A	FLVCR1	1	212885292	not provided	
NM_020247.4(COQ8A):c.1228C>T (p.Arg410Ter)	COQ8A	1	226983826	not provided	
NM_006015.4(ARID1A):c.1390C>T (p.Gln464Ter)	ARID1A	1	26731191	not provided	
NM_000350.2(ABCA4):c.5898+1G>A	ABCA4	1	94008234	not provided	
NM_001128425.1(MUTYH):c.463-1G>A	MUTYH	1	45332960	not provided	
NM_006516.2(SLC2A1):c.493G>A (p.Val165Ile)	SLC2A1	1	42930649	not provided	
NM_006516.2(SLC2A1):c.844C>T (p.Gln282Ter)	SLC2A1	1	42929616	not provided	
NM_000969.4(RPL5):c.535C>T (p.Arg179Ter)	RPL5	1	92837463	not provided	
NM_001165963.1(SCN1A):c.2946+5G>A	SCN1A	2	166037771	not provided	
NM_001165963.1(SCN1A):c.1170+1G>A	SCN1A	2	166047626	not provided	
NM_021007.2(SCN2A):c.4904G>A (p.Arg1635Gln)	SCN2A	2	165388710	not provided	
NM_000090.3(COL3A1):c.811C>T (p.Arg271Ter)	COL3A1	2	188991016	not provided	
NM_000090.3(COL3A1):c.1106G>A (p.Gly369Glu)	COL3A1	2	188993416	not provided	
NM_006922.3(SCN3A):c.3998C>T (p.Pro1333Leu)	SCN3A	2	165097493	not provided	
NM_021007.2(SCN2A):c.781G>A (p.Val261Met)	SCN2A	2	165310406	not provided	
NM_001165963.1(SCN1A):c.235G>A (p.Asp79Asn)	SCN1A	2	166073387	not provided	
NM_015265.3(SATB2):c.1165C>T (p.Arg389Cys)	SATB2	2	199348709	not provided	
NM_001256850.1(TTN):c.71942G>A (p.Trp23981Ter)	TTN	2	178569267	not provided	
NM_033084.4(FANCD2):c.990-1G>A	FANCD2	3	10043483	not provided	
NM_080916.2(DGUOK):c.443+1G>A	DGUOK	2	73946907	not provided	
NM_006302.2(MOGS):c.1801C>T (p.Arg601Ter)	MOGS	2	74461988	not provided	
NM_053025.3(MYLK):c.2119C>T (p.Gln707Ter)	MYLK	3	123708719	not provided	
NM_000094.3(COL7A1):c.7219G>A (p.Gly2407Ser)	COL7A1	3	48570914	not provided	
NM_000094.3(COL7A1):c.6109G>A (p.Gly2037Arg)	COL7A1	3	48575410	not provided	
NM_000094.3(COL7A1):c.4928G>A (p.Gly1643Asp)	COL7A1	3	48581129	not provided	
NM_003722.4(TP63):c.547C>T (p.Gln183Ter)	TP63	3	189808494	not provided	

NM_032682.5(FOXP1):c.574C>T (p.Gln192Ter)	FOXP1	3	71047032	not provided	
NM_014159.6(SETD2):c.5218C>T (p.Arg1740Trp)	SETD2	3	47088172	not provided	
NM_000094.3(COL7A1):c.6751G>A (p.Gly2251Arg)	COL7A1	3	48572942	not provided	
NM_000094.3(COL7A1):c.6082G>A (p.Gly2028Arg)	COL7A1	3	48575437	not provided	
NM_006005.3(WFS1):c.2170C>T (p.Pro724Ser)	WFS1	4	6301965	not provided	
NM_000248.3(MITF):c.909G>A (p.Thr303=)	MITF	3	69964897	not provided	
NM_032682.5(FOXP1):c.1489C>T (p.Arg497Ter)	FOXP1	3	70976982	not provided	
NM_001615.3(ACTG2):c.584C>T (p.Thr195Ile)	ACTG2	2	73913617	not provided	
NM_006846.3(SPINK5):c.2667-1G>A	SPINK5	5	148124764	not provided	
NM_014382.3(ATP2C1):c.2197C>T (p.Gln733Ter)	ATP2C1	3	130996750	not provided	
NM_005219.4(DIAPH1):c.2794C>T (p.Arg932Ter)	DIAPH1	5	141528926	not provided	
NM_000060.4(BTD):c.866C>T (p.Ala289Val)	BTD	3	15644722	not provided	
NM_003722.4(TP63):c.935G>A (p.Cys312Tyr)	TP63	3	189867885	not provided	
NM_003060.3(SLC22A5):c.77G>A (p.Ser26Asn)	SLC22A5	5	132370049	not provided	
NM_004100.4(EYA4):c.1537C>T (p.Gln513Ter)	EYA4	6	133515356	not provided	
NM_017651.4(AHI1):c.2609G>A (p.Trp870Ter)	AHI1	6	135428643	not provided	
NM_022455.4(NSD1):c.4765+1G>A	NSD1	5	177251854	not provided	
NM_000404.3(GLB1):c.902C>T (p.Ala301Val)	GLB1	3	33051895	not provided	
NM_023073.3(C5orf42):c.1270C>T (p.Arg424Ter)	C5orf42	5	37227669	not provided	
NM_032861.3(SERAC1):c.493C>T (p.Gln165Ter)	SERAC1	6	158144415	not provided	
NM_181523.2(PIK3R1):c.1042C>T (p.Arg348Ter)	PIK3R1	5	68293123	not provided	
NM_000046.3(ARSB):c.1325C>T (p.Thr442Met)	ARSB	5	78781863	not provided	
NM_001080.3(ALDH5A1):c.526G>A (p.Gly176Arg)	ALDH5A1	6	24503350	not provided	
NM_022455.4(NSD1):c.6010-1G>A	NSD1	5	177283786	not provided	
NM_000094.3(COL7A1):c.4670G>A (p.Gly1557Glu)	COL7A1	3	48581758	not provided	
NM_001723.5(DST):c.7543C>T (p.Gln2515Ter)	DST	6	56615924	not provided	
NM_001723.5(DST):c.4993C>T (p.Gln1665Ter)	DST	6	56619041	not provided	
NM_178012.4(TUBB2B):c.533C>T (p.Thr178Met)	TUBB2B	6	3225556	not provided	
NM_033071.3(SYNE1):c.18469C>T (p.Gln6157Ter)	SYNE1	6	152269178	not provided	
NM_001024630.3(RUNX2):c.569G>A (p.Arg190Gln)	RUNX2	6	45432008	not provided	
NM_000441.1(SLC26A4):c.2206C>T (p.Gln736Ter)	SLC26A4	7	107710170	not provided	
NM_000535.6(PMS2):c.2140C>T (p.Gln714Ter)	PMS2	7	5982858	not provided	
NM_022726.3(ELOVL4):c.506G>A (p.Trp169Ter)	ELOVL4	6	79921660	not provided	
NM_006766.4(KAT6A):c.4381C>T (p.Gln1461Ter)	KAT6A	8	41933839	not provided	
NM_014141.5(CNTNAP2):c.2050C>T (p.Gln684Ter)	CNTNAP2	7	147639258	not provided	
NM_153033.4(KCTD7):c.631C>T (p.Arg211Ter)	KCTD7	7	66638993	not provided	
NM_000238.3(KCNH2):c.1128+1810C>T	KCNH2	7	150955481	not provided	
NM_000089.3(COL1A2):c.407G>A (p.Gly136Asp)	COL1A2	7	94404867	not provided	
NM_003165.3(STXBP1):c.794+5G>A	STXBP1	9	127666301	not provided	
NM_017780.3(CHD7):c.1123C>T (p.Gln375Ter)	CHD7	8	60742555	not provided	
NM_003172.3(SURF1):c.681G>A (p.Trp227Ter)	SURF1	9	133352516	not provided	
NM_000426.3(LAMA2):c.283+1G>A	LAMA2	6	129050089	not provided	
NM_017890.4(VPS13B):c.2788C>T (p.Gln930Ter)	VPS13B	8	99275218	not provided	
NM_003165.3(STXBP1):c.1689G>A (p.Trp563Ter)	STXBP1	9	127682547	not provided	
NM_139025.4(ADAMTS13):c.3044+1G>A	ADAMTS13	9	133449966	not provided	

NM_004523.3(KIF11):c.77+1G>A	KIF11	10	92593453	not provided	
NM_000790.3(DDC):c.1073G>A (p.Arg358His)	DDC	7	50470140	not provided	
NM_024757.4(EHMT1):c.2622G>A (p.Trp874Ter)	EHMT1	9	137800894	not provided	
NM_052968.4(APOA5):c.289C>T (p.Gln97Ter)	APOA5	11	116790940	not provided	
NM_203447.3(DOCK8):c.2971-1G>A	DOCK8	9	396784	not provided	
NM_033056.3(PCDH15):c.1997+1G>A	PCDH15	10	54089983	not provided	
NM_000280.4(PAX6):c.486G>A (p.Trp162Ter)	PAX6	11	31800728	not provided	
NM_000543.4(SMPD1):c.1264-1G>A	SMPD1	11	6393616	not provided	
NM_000494.3(COL17A1):c.1906G>A (p.Gly636Arg)	COL17A1	10	104053664	not provided	
NM_000545.6(HNF1A):c.511C>T (p.Arg171Ter)	HNF1A	12	120989017	not provided	
NM_000545.6(HNF1A):c.686G>A (p.Arg229Gln)	HNF1A	12	120993679	not provided	
NM_017890.4(VPS13B):c.292-1G>A	VPS13B	8	99096311	not provided	
NM_001197104.1(KMT2A):c.4504C>T (p.Arg1502Ter)	KMT2A	11	118489816	not provided	
NM_004572.3(PKP2):c.1147C>T (p.Gln383Ter)	PKP2	12	32868950	not provided	
NM_130799.2(MEN1):c.781C>T (p.Gln261Ter)	MEN1	11	64807554	not provided	
NM_024678.5(NARS2):c.727C>T (p.Arg243Ter)	NARS2	11	78493158	not provided	
NM_000372.4(TYR):c.976C>T (p.Gln326Ter)	TYR	11	89191358	not provided	
NM_000785.3(CYP27B1):c.305G>A (p.Gly102Glu)	CYP27B1	12	57766088	not provided	
NM_020661.3(AICDA):c.334C>T (p.Arg112Cys)	AICDA	12	8605308	not provided	
NM_002336.2(LRP6):c.3178C>T (p.Arg1060Ter)	LRP6	12	12148970	not provided	
NM_001845.5(COL4A1):c.2317G>A (p.Gly773Arg)	COL4A1	13	110179298	not provided	
NM_006755.1(TALDO1):c.574C>T (p.Arg192Cys)	TALDO1	11	763456	not provided	
NM_000059.3(BRCA2):c.4354C>T (p.Gln1452Ter)	BRCA2	13	32338709	not provided	
NM_170665.3(ATP2A2):c.1185-1G>A	ATP2A2	12	110333180	not provided	
NM_000545.6(HNF1A):c.788G>A (p.Arg263His)	HNF1A	12	120994238	not provided	
NM_000834.3(GRIN2B):c.1555C>T (p.Arg519Ter)	GRIN2B	12	13615213	not provided	
NG_017041.1:g.5147G>A	RMRP	9	35657872	not provided	
NM_013382.5(POMT2):c.1762C>T (p.Arg588Ter)	POMT2	14	77280044	not provided	
NM_014319.4(LEMD3):c.1873C>T (p.Arg625Ter)	LEMD3	12	65238766	not provided	
NM_000494.3(COL17A1):c.332-1G>A	COL17A1	10	104074232	not provided	
NM_001844.4(COL2A1):c.2818C>T (p.Arg940Ter)	COL2A1	12	47978674	not provided	
NM_003482.3(KMT2D):c.16522-1G>A	KMT2D	12	49021873	not provided	
NM_003482.3(KMT2D):c.2317C>T (p.Gln773Ter)	KMT2D	12	49051366	not provided	
NM_001005463.2(EBF3):c.370C>T (p.Gln124Ter)	EBF3	10	129962212	not provided	
NM_052867.3(NALCN):c.2008C>T (p.Gln670Ter)	NALCN	13	101143190	not provided	
NM_000520.5(HEXA):c.748G>A (p.Gly250Ser)	HEXA	15	72350575	not provided	
NM_000257.3(MYH7):c.5655G>A (p.Ala1885=)	MYH7	14	23414007	not provided	
NM_015087.4(SPG20):c.1369C>T (p.Arg457Ter)	SPG20	13	36314341	not provided	
NM_000051.3(ATM):c.8641C>T (p.Gln2881Ter)	ATM	11	108347335	not provided	
NM_001271.3(CHD2):c.4999C>T (p.Gln1667Ter)	CHD2	15	93020104	not provided	
NM_173201.3(ATP2A1):c.1184+1G>A	ATP2A1	16	28894244	not provided	
NM_000781.2(CYP11A1):c.358C>T (p.Arg120Ter)	CYP11A1	15	74347967	not provided	
NM_015272.4(RPGRIP1L):c.1700-1G>A	RPGRIP1L	16	53652988	not provided	
NM_015335.4(MED13L):c.4051C>T (p.Gln1351Ter)	MED13L	12	115987172	not provided	
NM_002693.2(POLG):c.2125C>T (p.Arg709Ter)	POLG	15	89323847	not provided	

NM_000382.2(ALDH3A2):c.1202G>A (p.Gly401Glu)	ALDH3A2	17	19665042	not provided	
NM_000430.3(PAFAH1B1):c.1159+1G>A	PAFAH1B1	17	2680321	not provided	
NM_001271.3(CHD2):c.1809+1G>A	CHD2	15	92955513	not provided	
NM_003632.2(CNTNAP1):c.1282C>T (p.Arg428Ter)	CNTNAP1	17	42687957	not provided	
NM_152641.3(ARID2):c.2872C>T (p.Gln958Ter)	ARID2	12	45850995	not provided	
NM_004247.3(EFTUD2):c.1732C>T (p.Arg578Ter)	EFTUD2	17	44860033	not provided	
NM_004380.2(CREBBP):c.2659C>T (p.Gln887Ter)	CREBBP	16	3770791	not provided	
NM_005682.6(ADGRG1):c.367C>T (p.Gln123Ter)	ADGRG1	16	57651502	not provided	
NM_004004.5(GJB2):c.23C>T (p.Thr8Met)	GJB2	13	20189559	not provided	
NM_013275.5(ANKRD11):c.7570-1G>A	ANKRD11	16	89274958	not provided	
NM_016239.3(MYO15A):c.9876G>A (p.Trp3292Ter)	MYO15A	17	18166449	not provided	
NM_020366.3(RPGRIP1):c.3565C>T (p.Arg1189Ter)	RPGRIP1	14	21345145	not provided	
NM_001170629.1(CHD8):c.1690C>T (p.Arg564Ter)	CHD8	14	21426154	not provided	
NM_144612.6(LOXHD1):c.4741-1G>A	LOXHD1	18	46524602	not provided	
NM_001163809.1(WDR81):c.1564C>T (p.Gln522Ter)	WDR81	17	1726523	not provided	
NM_001083962.1(TCF4):c.1727G>A (p.Arg576Gln)	TCF4	18	55228999	not provided	
NM_000458.3(HNF1B):c.1108G>A (p.Gly370Ser)	HNF1B	17	37710601	not provided	
NM_004247.3(EFTUD2):c.1096C>T (p.Gln366Ter)	EFTUD2	17	44867860	not provided	
NM_024884.2(L2HGDH):c.256+1G>A	L2HGDH	14	50302901	not provided	
NM_001127221.1(CACNA1A):c.4046G>A (p.Arg1349Gln)	CACNA1A	19	13262780	not provided	
NM_002734.4(PRKAR1A):c.535C>T (p.Gln179Ter)	PRKAR1A	17	68524944	not provided	
NM_017882.2(CLN6):c.167G>A (p.Trp56Ter)	CLN6	15	68218567	not provided	
NM_007254.3(PNKP):c.1386+1G>A	PNKP	19	49861607	not provided	
NM_000546.5(TP53):c.673-1G>A	TP53	17	7674291	not provided	
NM_002693.2(POLG):c.694C>T (p.Arg232Cys)	POLG	15	89330242	not provided	
NM_000022.3(ADA):c.7C>T (p.Gln3Ter)	ADA	20	44651601	not provided	
NM_000159.3(GCDH):c.395G>A (p.Arg132Gln)	GCDH	19	12893543	not provided	
NM_000159.3(GCDH):c.937C>T (p.Arg313Trp)	GCDH	19	12896994	not provided	
NM_173483.3(CYP4F22):c.1084C>T (p.Arg362Ter)	CYP4F22	19	15544227	not provided	
NM_001199107.1(TBC1D24):c.121C>T (p.Gln41Ter)	TBC1D24	16	2496269	not provided	
NM_004380.2(CREBBP):c.2302C>T (p.Arg768Ter)	CREBBP	16	3773912	not provided	
NM_016941.3(DLL3):c.661C>T (p.Arg221Ter)	DLL3	19	39504079	not provided	
NM_012213.2(MLYCD):c.946C>T (p.Gln316Ter)	MLYCD	16	83912365	not provided	
NM_015629.3(PRPF31):c.855+1G>A	PRPF31	19	54124657	not provided	
NM_000303.2(PMM2):c.157C>T (p.Gln53Ter)	PMM2	16	8801889	not provided	
NM_007254.3(PNKP):c.1315C>T (p.Arg439Ter)	PNKP	19	49861679	not provided	
NM_174917.4(ACSF3):c.1608G>A (p.Trp536Ter)	ACSF3	16	89146044	not provided	
NM_013275.5(ANKRD11):c.7138C>T (p.Gln2380Ter)	ANKRD11	16	89279404	not provided	
NM_006086.3(TUBB3):c.862G>A (p.Glu288Lys)	TUBB3	16	89935313	not provided	
NM_000833.4(GRIN2A):c.487C>T (p.Gln163Ter)	GRIN2A	16	9938479	not provided	
NM_000444.5(PHEX):c.58C>T (p.Arg20Ter)	PHEX	X	22033063	not provided	
NM_000167.5(GK):c.514C>T (p.Arg172Ter)	GK	X	30694499	not provided	
NM_004006.2(DMD):c.5758C>T (p.Gln1920Ter)	DMD	X	32342264	not provided	
NM_001039590.2(USP9X):c.643C>T (p.Arg215Ter)	USP9X	X	41137011	not provided	
NM_000516.5(GNAS):c.125G>A (p.Arg42His)	GNAS	20	58891851	not provided	

NM_014009.3(FOXP3):c.1190G>A (p.Arg397Gln)	FOXP3	X	49251440	not provided	
NM_001111125.2(IQSEC2):c.2507C>T (p.Ala836Val)	IQSEC2	X	53248189	not provided	
NM_000383.3(AIRE):c.274C>T (p.Arg92Trp)	AIRE	21	44286698	not provided	
NM_000088.3(COL1A1):c.1930G>A (p.Gly644Ser)	COL1A1	17	50192528	not provided	
NM_001849.3(COL6A2):c.1572+1G>A	COL6A2	21	46122159	not provided	
NM_152906.6(TANGO2):c.94C>T (p.Arg32Ter)	TANGO2	22	20043392	not provided	
NM_000018.3(ACADVL):c.1843C>T (p.Arg615Ter)	ACADVL	17	7224972	not provided	
NM_000152.4(GAA):c.1478C>T (p.Pro493Leu)	GAA	17	80110767	not provided	
NM_000444.5(PHEX):c.565C>T (p.Gln189Ter)	PHEX	X	22077604	not provided	
NM_000444.5(PHEX):c.849+1G>A	PHEX	X	22094100	not provided	
NM_003639.4(IKBKG):c.502C>T (p.Gln168Ter)	IKBKG	X	154558634	not provided	
NM_001363.4(DKC1):c.1133G>A (p.Arg378Gln)	DKC1	X	154773227	not provided	
NM_001145408.1(NONO):c.103C>T (p.Gln35Ter)	NONO	X	71290740	not provided	
NM_001111125.2(IQSEC2):c.2776C>T (p.Arg926Ter)	IQSEC2	X	53243445	not provided	
NM_031407.6(HUWE1):c.328C>T (p.Arg110Trp)	HUWE1	X	53647391	not provided	
NM_002547.2(OPHN1):c.1246C>T (p.Gln416Ter)	OPHN1	X	68192949	not provided	
NM_001399.4(EDA):c.617C>T (p.Pro206Leu)	EDA	X	70027947	not provided	
NM_000206.2(IL2RG):c.202G>A (p.Glu68Lys)	IL2RG	X	71110964	not provided	
NM_020436.4(SALL4):c.2491C>T (p.Arg831Ter)	SALL4	20	51789112	not provided	
NM_003560.3(PLA2G6):c.1501G>A (p.Glu501Lys)	PLA2G6	22	38123185	not provided	
NM_000132.3(F8):c.6103G>A (p.Val2035Met)	F8	X	154902063	not provided	
NM_000377.2(WAS):c.360+1G>A	WAS	X	48685634	not provided	
NM_013296.4(GPSM2):c.1093C>T (p.Arg365Ter)	GPSM2	1	108904155	not provided	
NM_002016.1(FLG):c.5392C>T (p.Arg1798Ter)	FLG	1	152309494	not provided	
NM_020699.3(GATAD2B):c.973C>T (p.Gln325Ter)	GATAD2B	1	153816516	not provided	
NM_198053.2(CD247):c.301C>T (p.Gln101Ter)	CD247	1	167435434	not provided	
NM_000228.2(LAMB3):c.241C>T (p.Arg81Ter)	LAMB3	1	209638591	not provided	
NM_206933.2(USH2A):c.8167C>T (p.Arg2723Ter)	USH2A	1	215888482	not provided	
NM_206933.2(USH2A):c.3507G>A (p.Trp1169Ter)	USH2A	1	216199931	not provided	
NM_021007.2(SCN2A):c.4303C>T (p.Arg1435Ter)	SCN2A	2	165377645	not provided	
NM_001256850.1(TTN):c.82117C>T (p.Arg27373Ter)	TTN	2	178558419	not provided	
NM_001256850.1(TTN):c.81440G>A (p.Trp27147Ter)	TTN	2	178559769	not provided	
NM_001256850.1(TTN):c.74680C>T (p.Gln24894Ter)	TTN	2	178566529	not provided	
NM_001256850.1(TTN):c.73481G>A (p.Trp24494Ter)	TTN	2	178567728	not provided	
NM_001256850.1(TTN):c.71355G>A (p.Trp23785Ter)	TTN	2	178569854	not provided	
NM_000090.3(COL3A1):c.3499G>A (p.Gly1167Ser)	COL3A1	2	189008116	not provided	
NM_014362.3(HIBCH):c.809+1G>A	HIBCH	2	190246153	not provided	
NM_032504.1(UNC80):c.66955G>A (p.Trp2232Ter)	UNC80	2	209941467	not provided	
NM_173076.2(ABCA12):c.7444C>T (p.Arg2482Ter)	ABCA12	2	214937608	not provided	
NM_000751.2(CHRND):c.127C>T (p.Arg43Trp)	CHRND	2	232526603	not provided	
NM_000463.2(UGT1A1):c.1084G>A (p.Gly362Ser)	UGT1A1	2	233767936	not provided	
NM_004369.3(COL6A3):c.2506C>T (p.Arg836Ter)	COL6A3	2	237377336	not provided	
NM_004321.7(KIF1A):c.920G>A (p.Arg307Gln)	KIF1A	2	240775889	not provided	
NM_175629.2(DNMT3A):c.2311C>T (p.Arg771Ter)	DNMT3A	2	25240313	not provided	
NM_014946.3(SPAST):c.1729-1G>A	SPAST	2	32154373	not provided	

NM_000179.2(MSH6):c.1714C>T (p.Gln572Ter)	MSH6	2	47799697	not provided	
NM_000179.2(MSH6):c.3140G>A (p.Trp1047Ter)	MSH6	2	47801123	not provided	
NM_003494.3(DYSF):c.5884C>T (p.Gln1962Ter)	DYSF	2	71679173	not provided	
NM_006463.4(STAMBP):c.938C>T (p.Thr313Ile)	STAMBP	2	73850446	not provided	
NM_000388.3(CASR):c.577C>T (p.Gln193Ter)	CASR	3	122261612	not provided	
NM_006218.3(PIK3CA):c.278G>A (p.Arg93Gln)	PIK3CA	3	179199103	not provided	
NM_006218.3(PIK3CA):c.1093G>A (p.Glu365Lys)	PIK3CA	3	179204536	not provided	
NM_000249.3(MLH1):c.2142G>A (p.Trp714Ter)	MLH1	3	37050524	not provided	
NM_001904.3(CTNNB1):c.1759C>T (p.Arg587Ter)	CTNNB1	3	41235799	not provided	
NM_000094.3(COL7A1):c.7828C>T (p.Arg2610Ter)	COL7A1	3	48568137	not provided	
NM_000094.3(COL7A1):c.7688G>A (p.Gly2563Asp)	COL7A1	3	48568854	not provided	
NM_000094.3(COL7A1):c.6900+1G>A	COL7A1	3	48572670	not provided	
NM_000094.3(COL7A1):c.3631C>T (p.Gln1211Ter)	COL7A1	3	48586166	not provided	
NM_004656.3(BAP1):c.1777C>T (p.Gln593Ter)	BAP1	3	52403251	not provided	
NM_024582.4(FAT4):c.2527C>T (p.Gln843Ter)	FAT4	4	125318938	not provided	
NM_000038.5(APC):c.583C>T (p.Gln195Ter)	APC	5	112780841	not provided	
NM_000038.5(APC):c.1778G>A (p.Trp593Ter)	APC	5	112834985	not provided	
NM_000038.5(APC):c.5764C>T (p.Gln1922Ter)	APC	5	112841358	not provided	
NM_001182.4(ALDH7A1):c.177G>A (p.Trp59Ter)	ALDH7A1	5	126595022	not provided	
NM_001135243.1(TCOF1):c.2140C>T (p.Gln714Ter)	TCOF1	5	150376328	not provided	
NM_000806.5(GABRA1):c.752G>A (p.Gly251Asp)	GABRA1	5	161890946	not provided	
NM_001130105.1(COL4A3BP):c.779C>T (p.Ser260Leu)	COL4A3BP	5	75426432	not provided	
NM_000426.3(LAMA2):c.5866-1G>A	LAMA2	6	129427751	not provided	
NM_000045.3(ARG1):c.703G>A (p.Gly235Arg)	ARG1	6	131583392	not provided	
NM_005670.3(EPM2A):c.163C>T (p.Gln55Ter)	EPM2A	6	145735336	not provided	
NM_020732.3(ARID1B):c.4456C>T (p.Gln1486Ter)	ARID1B	6	157201050	not provided	
NM_006772.2(SYNGAP1):c.3471G>A (p.Trp1157Ter)	SYNGAP1	6	33444506	not provided	
NM_004415.3(DSP):c.8014C>T (p.Gln2672Ter)	DSP	6	7585276	not provided	
NM_000238.3(KCNH2):c.1726C>T (p.Gln576Ter)	KCNH2	7	150951667	not provided	
NM_000238.3(KCNH2):c.472+1G>A	KCNH2	7	150959571	not provided	
NM_152743.3(BRAT1):c.1771-1G>A	BRAT1	7	2538765	not provided	
NM_003718.4(CDK13):c.2209C>T (p.Arg737Cys)	CDK13	7	40001887	not provided	
NM_000162.3(GCK):c.184G>A (p.Val62Met)	GCK	7	44153325	not provided	
NM_001101.3(ACTB):c.629G>A (p.Arg210His)	ACTB	7	5528454	not provided	
NM_000501.3(ELN):c.631C>T (p.Pro211Ser)	ELN	7	74046755	not provided	
NM_000089.3(COL1A2):c.1054G>A (p.Gly352Ser)	COL1A2	7	94410260	not provided	
NM_000089.3(COL1A2):c.1702G>A (p.Gly568Ser)	COL1A2	7	94414258	not provided	
NM_000089.3(COL1A2):c.2314G>A (p.Gly772Ser)	COL1A2	7	94421027	not provided	
NM_014112.4(TRPS1):c.2421G>A (p.Trp807Ter)	TRPS1	8	115587280	not provided	
NM_000127.2(EXT1):c.250C>T (p.Gln84Ter)	EXT1	8	118110797	not provided	
NM_006766.4(KAT6A):c.4645G>A (p.Gly1549Ser)	KAT6A	8	41933575	not provided	
NM_006766.4(KAT6A):c.931C>T (p.Arg311Ter)	KAT6A	8	41978754	not provided	
NM_017780.3(CHD7):c.7879C>T (p.Arg2627Ter)	CHD7	8	60862244	not provided	
NM_153704.5(TMEM67):c.8876G>A (p.Trp296Ter)	TMEM67	8	93780891	not provided	
NM_003165.3(STXBP1):c.84G>A (p.Trp28Ter)	STXBP1	9	127651649	not provided	

NM_003165.3(STXBP1):c.748C>T (p.Gln250Ter)	STXBP1	9	127666250	not provided	
NM_003165.3(STXBP1):c.889G>A (p.Ala297Thr)	STXBP1	9	127668174	not provided	
NM_007171.3(POMT1):c.1858C>T (p.Arg620Ter)	POMT1	9	131521439	not provided	
NM_000368.4(TSC1):c.107-1G>A	TSC1	9	132927305	not provided	
NM_007327.3(GRIN1):c.1955C>T (p.Ala652Val)	GRIN1	9	137162681	not provided	
NM_024757.4(EHMT1):c.3127C>T (p.Gln1043Ter)	EHMT1	9	137813477	not provided	
NM_175073.2(APTX):c.124C>T (p.Arg42Ter)	APTX	9	32989768	not provided	
NM_000155.3(GALT):c.899G>A (p.Trp300Ter)	GALT	9	34649076	not provided	
NM_001003800.1(BICD2):c.2231C>T (p.Ser744Phe)	BICD2	9	92717824	not provided	
NM_000136.2(FANCC):c.1069C>T (p.Gln357Ter)	FANCC	9	95117318	not provided	
NM_000264.3(PTCH1):c.2833C>T (p.Arg945Ter)	PTCH1	9	95459654	not provided	
NM_000264.3(PTCH1):c.2701C>T (p.Gln901Ter)	PTCH1	9	95461858	not provided	
NM_000264.3(PTCH1):c.590G>A (p.Trp197Ter)	PTCH1	9	95482198	not provided	
NM_018294.5(CWF19L1):c.622C>T (p.Arg208Ter)	CWF19L1	10	100253422	not provided	
NM_001005463.2(EBF3):c.907C>T (p.Arg303Ter)	EBF3	10	129867246	not provided	
NM_016628.4(WAC):c.1198C>T (p.Gln400Ter)	WAC	10	28610731	not provided	
NM_000429.2(MAT1A):c.895C>T (p.Arg299Cys)	MAT1A	10	80275073	not provided	
NM_004329.2(BMPR1A):c.730C>T (p.Arg244Ter)	BMPR1A	10	86917188	not provided	
NM_000314.6(PTEN):c.209+1G>A	PTEN	10	87925558	not provided	
NM_001080463.1(DYNC2H1):c.10219C>T (p.Arg3407Ter)	DYNC2H1	11	103253440	not provided	
NM_000051.3(ATM):c.8140C>T (p.Gln2714Ter)	ATM	11	108335098	not provided	
NM_024426.4(WT1):c.1385G>A (p.Arg462Gln)	WT1	11	32392019	not provided	
NM_000256.3(MYBPC3):c.1621C>T (p.Gln541Ter)	MYBPC3	11	47342581	not provided	
NM_130799.2(MEN1):c.446-1G>A	MEN1	11	64808100	not provided	
NM_032193.3(RNASEH2C):c.451C>T (p.Pro151Ser)	RNASEH2C	11	65720062	not provided	
NM_032193.3(RNASEH2C):c.450G>A (p.Trp150Ter)	RNASEH2C	11	65720063	not provided	
NM_007103.3(NDUFV1):c.1156C>T (p.Arg386Cys)	NDUFV1	11	67611972	not provided	
NM_015531.5(C2CD3):c.3910+1G>A	C2CD3	11	74085617	not provided	
NM_012193.3(FZD4):c.611G>A (p.Cys204Tyr)	FZD4	11	86952145	not provided	
NM_170665.3(ATP2A2):c.1429C>T (p.Gln477Ter)	ATP2A2	12	110339290	not provided	
NM_000192.3(TBX5):c.1078C>T (p.Gln360Ter)	TBX5	12	114356011	not provided	
NM_015335.4(MED13L):c.5589-1G>A	MED13L	12	115975314	not provided	
NM_015335.4(MED13L):c.2524C>T (p.Arg842Ter)	MED13L	12	116003048	not provided	
NM_000545.6(HNF1A):c.685C>T (p.Arg229Ter)	HNF1A	12	120993678	not provided	
NM_001844.4(COL2A1):c.2491G>A (p.Gly831Arg)	COL2A1	12	47980941	not provided	
NM_001844.4(COL2A1):c.1330G>A (p.Gly444Ser)	COL2A1	12	47987113	not provided	
NM_003482.3(KMT2D):c.4148G>A (p.Cys1383Tyr)	KMT2D	12	49048053	not provided	
NM_006009.3(TUBA1A):c.1168C>T (p.Arg390Cys)	TUBA1A	12	49185198	not provided	
NM_006009.3(TUBA1A):c.190C>T (p.Arg64Trp)	TUBA1A	12	49186647	not provided	
NM_000020.2(ACVRL1):c.1027C>T (p.Gln343Ter)	ACVRL1	12	51915479	not provided	
NM_001131025.1(PEX5):c.552-1G>A	PEX5	12	7201750	not provided	
NM_025114.3(CEP290):c.3461+1G>A	CEP290	12	88092680	not provided	
NM_000131.4(F7):c.1091G>A (p.Arg364Gln)	F7	13	113118698	not provided	
NM_000321.2(RB1):c.607+1G>A	RB1	13	48349024	not provided	

NM_024570.3(RNASEH2B):c.437-1G>A	RNASEH2B	13	50943320	not provided	
NM_001170629.1(CHD8):c.4414C>T (p.Arg1472Ter)	CHD8	14	21400569	not provided	
NM_000359.2(TGM1):c.788G>A (p.Trp263Ter)	TGM1	14	24260028	not provided	
NM_001244189.1(KIAA0586):c.946C>T (p.Gln316Ter)	KIAA0586	14	58444155	not provided	
NM_001244189.1(KIAA0586):c.2512C>T (p.Arg838Ter)	KIAA0586	14	58467833	not provided	
NM_005982.3(SIX1):c.329G>A (p.Arg110Gln)	SIX1	14	60648861	not provided	
NM_130838.1(UBE3A):c.1516C>T (p.Arg506Cys)	UBE3A	15	25370598	not provided	
NM_014080.4(DUOX2):c.3329G>A (p.Arg1110Gln)	DUOX2	15	45099748	not provided	
NM_000138.4(FBN1):c.6616+1G>A	FBN1	15	48434593	not provided	
NM_000138.4(FBN1):c.6037+1G>A	FBN1	15	48444540	not provided	
NM_005902.3(SMAD3):c.1102C>T (p.Arg368Ter)	SMAD3	15	67187457	not provided	
NM_001145358.1(SIN3A):c.2689C>T (p.Arg897Ter)	SIN3A	15	75392404	not provided	
NM_001171.5(ABCC6):c.4081G>A (p.Asp1361Asn)	ABCC6	16	16154755	not provided	
NM_001171.5(ABCC6):c.2359G>A (p.Val787Ile)	ABCC6	16	16178854	not provided	
NM_000548.4(TSC2):c.328C>T (p.Gln110Ter)	TSC2	16	2053444	not provided	
NM_000548.4(TSC2):c.2221-1G>A	TSC2	16	2072848	not provided	
NM_000548.4(TSC2):c.4936G>A (p.Val1646Met)	TSC2	16	2086818	not provided	
NM_001009944.2(PKD1):c.6913C>T (p.Gln2305Ter)	PKD1	16	2108254	not provided	
NM_001009944.2(PKD1):c.1687C>T (p.Gln563Ter)	PKD1	16	2116564	not provided	
NM_024675.3(PALB2):c.163C>T (p.Gln55Ter)	PALB2	16	23637898	not provided	
NM_001199107.1(TBC1D24):c.58C>T (p.Gln20Ter)	TBC1D24	16	2496206	not provided	
NM_020988.2(GNAO1):c.709G>A (p.Glu237Lys)	GNAO1	16	56336846	not provided	
NM_016373.3(WWOX):c.1115G>A (p.Gly372Glu)	WWOX	16	79211666	not provided	
NM_016239.3(MYO15A):c.4032+1G>A	MYO15A	17	18127166	not provided	
NM_016239.3(MYO15A):c.4519C>T (p.Arg1507Ter)	MYO15A	17	18135747	not provided	
NM_000267.3(NF1):c.801G>A (p.Trp267Ter)	NF1	17	31182578	not provided	
NM_000526.4(KRT14):c.1163G>A (p.Arg388His)	KRT14	17	41583346	not provided	
NM_004247.3(EFTUD2):c.2466+1G>A	EFTUD2	17	44853516	not provided	
NM_000088.3(COL1A1):c.3416G>A (p.Gly1139Asp)	COL1A1	17	50187491	not provided	
NM_032043.2(BRIP1):c.2493-1G>A	BRIP1	17	61693513	not provided	
NM_032043.2(BRIP1):c.2258-1G>A	BRIP1	17	61743135	not provided	
NM_000789.3(ACE):c.412C>T (p.Gln138Ter)	ACE	17	63478093	not provided	
NM_000546.5(TP53):c.836G>A (p.Gly279Glu)	TP53	17	7673784	not provided	
NM_000546.5(TP53):c.159G>A (p.Trp53Ter)	TP53	17	7676210	not provided	
NM_000271.4(NPC1):c.1298C>T (p.Pro433Leu)	NPC1	18	23556271	not provided	
NM_030632.2(ASXL3):c.3364C>T (p.Gln1122Ter)	ASXL3	18	33743212	not provided	
NM_030632.2(ASXL3):c.4360C>T (p.Gln1454Ter)	ASXL3	18	33744208	not provided	
NM_001083962.1(TCF4):c.652C>T (p.Gln218Ter)	TCF4	18	55279554	not provided	
NM_001128849.1(SMARCA4):c.2654G>A (p.Arg885His)	SMARCA4	19	11021762	not provided	
NM_000527.4(LDLR):c.1439C>T (p.Ala480Val)	LDLR	19	11113615	not provided	
NM_001127221.1(CACNA1A):c.5396C>T (p.Ser1799Leu)	CACNA1A	19	13231717	not provided	
NM_001127221.1(CACNA1A):c.4058C>T (p.Pro1353Leu)	CACNA1A	19	13262768	not provided	
NM_173483.3(CYP4F22):c.976C>T (p.Arg326Ter)	CYP4F22	19	15544007	not provided	
NM_000540.2(RYR1):c.4672C>T (p.Gln1558Ter)	RYR1	19	38483078	not provided	
NM_000540.2(RYR1):c.14512-1G>A	RYR1	19	38580369	not provided	

NM_020533.2(MCOLN1):c.405+1G>A	MCOLN1	19	7526607	not provided	
NM_000214.2(JAG1):c.863G>A (p.Trp288Ter)	JAG1	20	10652491	not provided	
NM_032195.2(SON):c.3334C>T (p.Arg1112Ter)	SON	21	33552565	not provided	
NM_001396.4(DYRK1A):c.517-1G>A	DYRK1A	21	37486466	not provided	
NM_001396.4(DYRK1A):c.691C>T (p.Arg231Ter)	DYRK1A	21	37490201	not provided	
NM_001396.4(DYRK1A):c.1546+1G>A	DYRK1A	21	37505590	not provided	
NM_000211.4(ITGB2):c.2013G>A (p.Trp671Ter)	ITGB2	21	44888760	not provided	
NM_001848.2(COL6A1):c.815G>A (p.Gly272Asp)	COL6A1	21	45989094	not provided	
NM_007194.3(CHEK2):c.28C>T (p.Gln10Ter)	CHEK2	22	28734694	not provided	
NM_000268.3(NF2):c.517-1G>A	NF2	22	29655593	not provided	
NM_001429.3(EP300):c.319C>T (p.Gln107Ter)	EP300	22	41117411	not provided	
NM_000061.2(BTK):c.1185G>A (p.Trp395Ter)	BTK	X	101356948	not provided	
NM_000061.2(BTK):c.894+1G>A	BTK	X	101359292	not provided	
NM_178153.2(DCX):c.505C>T (p.Gln169Ter)	DCX	X	111401190	not provided	
NM_000033.3(ABCD1):c.1895C>T (p.Thr632Ile)	ABCD1	X	153743250	not provided	
NM_000425.4(L1CAM):c.1939+1G>A	L1CAM	X	153867799	not provided	
NM_000425.4(L1CAM):c.1408C>T (p.Gln470Ter)	L1CAM	X	153868699	not provided	
NM_000284.3(PDHA1):c.905G>A (p.Arg302His)	PDHA1	X	19358921	not provided	
NM_014927.4(CNKSR2):c.1282C>T (p.Arg428Ter)	CNKSR2	X	21532046	not provided	
NM_014927.4(CNKSR2):c.2145+1G>A	CNKSR2	X	21666880	not provided	
NM_000444.5(PHEX):c.151C>T (p.Gln51Ter)	PHEX	X	22038501	not provided	
NM_000444.5(PHEX):c.670C>T (p.Gln224Ter)	PHEX	X	22090435	not provided	
NM_000444.5(PHEX):c.1147C>T (p.Gln383Ter)	PHEX	X	22111534	not provided	
NM_004006.2(DMD):c.10651C>T (p.Gln3551Ter)	DMD	X	31147421	not provided	
NM_004006.2(DMD):c.8390+1G>A	DMD	X	31507280	not provided	
NM_004006.2(DMD):c.3487C>T (p.Gln1163Ter)	DMD	X	32454778	not provided	
NM_004006.2(DMD):c.2555G>A (p.Trp852Ter)	DMD	X	32491344	not provided	
NM_001039590.2(USP9X):c.7207C>T (p.Gln2403Ter)	USP9X	X	41229398	not provided	
NM_001193416.2(DDX3X):c.121C>T (p.Pro41Ser)	DDX3X	X	41339053	not provided	
NM_003688.3(CASK):c.1837C>T (p.Arg613Ter)	CASK	X	41555605	not provided	
NM_003688.3(CASK):c.1480C>T (p.Gln494Ter)	CASK	X	41578363	not provided	
NM_000377.2(WAS):c.273+1G>A	WAS	X	48684424	not provided	
NM_000377.2(WAS):c.1453G>A (p.Asp485Asn)	WAS	X	48689434	not provided	
NM_005710.2(PQBP1):c.641+1G>A	PQBP1	X	48902796	not provided	
NM_001111125.2(IQSEC2):c.3250C>T (p.Gln1084Ter)	IQSEC2	X	53238172	not provided	
NM_000044.4(AR):c.2420G>A (p.Cys807Tyr)	AR	X	67721934	not provided	
NM_000044.4(AR):c.2450-1G>A	AR	X	67722826	not provided	
NM_000206.2(IL2RG):c.545G>A (p.Cys182Tyr)	IL2RG	X	71110205	not provided	
NM_001008537.2(KIAA2022):c.766C>T (p.Gln256Ter)	KIAA2022	X	74743791	not provided	
NM_032121.5(MAGT1):c.110G>A (p.Trp37Ter)	MAGT1	X	77895397	not provided	
NM_000552.4(VWF):c.3614G>A (p.Arg1205His)	VWF	12	6021960	not provided;von Willebrand disease type 1	
NM_000552.4(VWF):c.2279G>A (p.Arg760His)	VWF	12	6046725	not provided;von Willebrand disease type 1	
NM_000552.4(VWF):c.2561G>A (p.Arg854Gln)	VWF	12	6034812	not provided;von Willebrand disease type 1;von Willebrand disease type 2;von Willebrand disease type 2N;von Willebrand disorder	
NM_000552.4(VWF):c.4975C>T (p.Arg1659Ter)	VWF	12	6018443	not provided;von Willebrand disease type 1;von Willebrand disease type 3	

NM_000552.4(VWF):c.5557C>T (p.Arg1853Ter)	VWF	12	6013544	not provided;von Willebrand disease type 1;von Willebrand disease type 3
NM_000552.4(VWF):c.3970G>A (p.Gly1324Ser)	VWF	12	6019448	not provided;von Willebrand disease type 2M
NM_000552.4(VWF):c.3854C>T (p.Ser1285Phe)	VWF	12	6019564	not provided;von Willebrand disease type 2M
NM_000552.4(VWF):c.2372C>T (p.Thr791Met)	VWF	12	6044361	not provided;von Willebrand disease type 2N
NM_000552.4(VWF):c.2446C>T (p.Arg816Trp)	VWF	12	6036488	not provided;von Willebrand disease type 2N
NM_000552.4(VWF):c.7603C>T (p.Arg2535Ter)	VWF	12	5969337	not provided;von Willebrand disease type 3
NM_000552.4(VWF):c.4789C>T (p.Arg1597Trp)	VWF	12	6018629	not provided;von Willebrand disease, type 2a
NM_000552.4(VWF):c.1648G>A (p.Gly550Arg)	VWF	12	6057930	not provided;von Willebrand disease, type 2a
NM_000552.4(VWF):c.3916C>T (p.Arg1306Trp)	VWF	12	6019502	not provided;von Willebrand disease, type 2b
NM_000552.4(VWF):c.3922C>T (p.Arg1308Cys)	VWF	12	6019496	not provided;von Willebrand disease, type 2b
NM_000552.4(VWF):c.3946G>A (p.Val1316Met)	VWF	12	6019472	not provided;von Willebrand disease, type 2b
NM_000552.4(VWF):c.4022G>A (p.Arg1341Gln)	VWF	12	6019396	not provided;von Willebrand disease, type 2b
NM_000518.4(HBB):c.209G>A (p.Gly70Asp)	HBB	11	5226683	not specified
NM_000518.4(HBB):c.22G>A (p.Glu8Lys)	HBB	11	5227000	not specified
NM_003491.3(NAA10):c.346C>T (p.Arg116Trp)	NAA10	X	153932111	N-terminal acetyltransferase deficiency;not provided
NM_001040613.2(TMEM70):c.238C>T (p.Arg80Ter)	TMEM70	8	73978783	Nuclearly-encoded mitochondrial complex V (ATP synthase) deficiency 2
NM_000273.2(GPR143):c.266C>T (p.Ser89Phe)	GPR143	X	9760811	Nystagmus 6, congenital, X-linked
NM_001134944.1(GHRL):c.116G>A (p.Arg39Gln)	GHRL	3	10289835	Obesity
NM_005912.2(MC4R):c.148G>A (p.Val50Met)	MC4R	18	60372202	Obesity
NM_005912.2(MC4R):c.812G>A (p.Cys271Tyr)	MC4R	18	60371538	Obesity
NM_005912.2(MC4R):c.523G>A (p.Ala175Thr)	MC4R	18	60371827	Obesity
NM_005912.2(MC4R):c.656C>T (p.Ala219Val)	MC4R	18	60371694	Obesity
NM_021969.2(NR0B2):c.100C>T (p.Arg34Ter)	NR0B2	1	26913841	Obesity, mild, early-onset
NM_003356.3(UCP3):c.427C>T (p.Arg143Ter)	UCP3	11	74005844	Obesity, severe, and type II diabetes
NM_003356.3(UCP3):c.208C>T (p.Arg70Trp)	UCP3	11	74006298	Obesity, severe, and type II diabetes
NM_178857.5(RP1L1):c.133C>T (p.Arg45Trp)	RP1L1	8	10623069	Occult macular dystrophy
NM_021828.4(HPSE2):c.1516C>T (p.Arg506Ter)	HPSE2	10	98482733	Ochoa syndrome
NM_021828.4(HPSE2):c.1414C>T (p.Arg472Ter)	HPSE2	10	98490103	Ochoa syndrome
NM_021828.4(HPSE2):c.457C>T (p.Arg153Ter)	HPSE2	10	99144391	Ochoa syndrome
NM_000273.2(GPR143):c.455G>A (p.Ser152Asn)	GPR143	X	9759332	Ocular albinism, type I;not provided
NM_000273.2(GPR143):c.104G>A (p.Gly35Asp)	GPR143	X	9765714	Ocular albinism, type I;not provided
NM_000372.4(TYR):c.1265G>A (p.Arg422Gln)	TYR	11	89284853	Oculocutaneous albinism type 1, temperature sensitive;not provided
NM_000372.4(TYR):c.242C>T (p.Pro81Leu)	TYR	11	89178195	Oculocutaneous albinism type 1B;Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.1147G>A (p.Asp383Asn)	TYR	11	89227933	Oculocutaneous albinism type 1B;Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.140G>A (p.Gly47Asp)	TYR	11	89178093	Oculocutaneous albinism type 1B;Tyrosinase-negative oculocutaneous albinism;not provided
NM_000550.2(TYRP1):c.1120C>T (p.Arg374Ter)	TYRP1	9	12704564	Oculocutaneous albinism type 3
NM_000550.2(TYRP1):c.1067G>A (p.Arg356Gln)	TYRP1	9	12702424	Oculocutaneous albinism type 3
NM_016180.4(SLC45A2):c.563-1G>A	SLC45A2	5	33964017	Oculocutaneous albinism type 4
NM_016180.4(SLC45A2):c.1457C>T (p.Ala486Val)	SLC45A2	5	33944784	Oculocutaneous albinism type 4
NM_016180.4(SLC45A2):c.469G>A (p.Asp157Asn)	SLC45A2	5	33982329	Oculocutaneous albinism type 4
NM_016180.4(SLC45A2):c.856C>T (p.Gln286Ter)	SLC45A2	5	33963723	Oculocutaneous albinism type 4
NM_000372.4(TYR):c.1217C>T (p.Pro406Leu)	TYR	11	89284805	Oculocutaneous albinism;Oculocutaneous albinism type 1B;not provided
NM_000275.2(OCA2):c.1327G>A (p.Val443Ile)	OCA2	15	27985101	Oculocutaneous albinism;Skin/hair/eye pigmentation, variation in, 1;Tyrosinase-positive oculocutaneous albinism;not provided
NM_000165.4(GJA1):c.61G>A (p.Gly21Arg)	GJA1	6	121446908	Oculodentodigital dysplasia

NM_000165.4(GJA1):c.65G>A (p.Gly22Glu)	GJA1	6	121446912	Oculodentodigital dysplasia
NM_000165.4(GJA1):c.286G>A (p.Val96Met)	GJA1	6	121447133	Oculodentodigital dysplasia
NM_000165.4(GJA1):c.31C>T (p.Leu11Phe)	GJA1	6	121446878	Oculodentodigital dysplasia
NM_000165.4(GJA1):c.97C>T (p.Arg33Ter)	GJA1	6	121446944	Oculodentodigital dysplasia, autosomal recessive
NM_000165.4(GJA1):c.227G>A (p.Arg76His)	GJA1	6	121447074	Oculodentodigital dysplasia, autosomal recessive
NM_017745.5(BCOR):c.254C>T (p.Pro85Leu)	BCOR	X	40075092	Oculofaciocardiodental syndrome
NM_001123383.1(BCOR):c.2926C>T (p.Arg976Ter)	BCOR	X	40072420	Oculofaciocardiodental syndrome
NM_022068.3(PIEZO2):c.7067C>T (p.Thr2356Met)	PIEZO2	18	10689746	Oculomelic amyoplasia
NM_022068.3(PIEZO2):c.6668C>T (p.Ser2223Leu)	PIEZO2	18	10696257	Oculomelic amyoplasia
NM_022068.3(PIEZO2):c.6662C>T (p.Thr2221Ile)	PIEZO2	18	10696263	Oculomelic amyoplasia
NM_000478.5(ALPL):c.323C>T (p.Pro108Leu)	ALPL	1	21563135	Odontohypophosphatasia
NM_025216.2(WNT10A):c.27G>A (p.Trp9Ter)	WNT10A	2	218881022	Odontoonychodermal dysplasia
NM_025216.2(WNT10A):c.383G>A (p.Arg128Gln)	WNT10A	2	218889990	Odontoonychodermal dysplasia;Tooth agenesis, selective, 4;not provided
NM_032208.2(ANTXR1):c.505C>T (p.Arg169Ter)	ANTXR1	2	69075602	Odontotrichomelic syndrome
NM_032208.2(ANTXR1):c.262C>T (p.Arg88Ter)	ANTXR1	2	69044779	Odontotrichomelic syndrome
NM_000541.4(SAG):c.577C>T (p.Arg193Ter)	SAG	2	233328542	Oguchi's disease
NM_000541.4(SAG):c.874C>T (p.Arg292Ter)	SAG	2	233335029	Oguchi's disease
NM_000541.4(SAG):c.523C>T (p.Arg175Ter)	SAG	2	233328488	Oguchi's disease
NM_005120.2(MED12):c.3443G>A (p.Arg1148His)	MED12	X	71128686	Ohdo syndrome, X-linked
NM_000088.3(COL1A1):c.572G>A (p.Gly191Asp)	COL1A1	17	50198177	Oi/eds combined syndrome
NM_000616.4(CD4):c.793C>T (p.Arg265Trp)	CD4	12	6816241	Okt4 epitope deficiency
NM_177559.2(CSNK2A1):c.140G>A (p.Arg47Gln)	CSNK2A1	20	505191	Okur-chung neurodevelopmental syndrome
NM_004655.3(AXIN2):c.1966C>T (p.Arg656Ter)	AXIN2	17	65536495	Oligodontia-colorectal cancer syndrome
NM_004655.3(AXIN2):c.1989G>A (p.Trp663Ter)	AXIN2	17	65536472	Oligodontia-colorectal cancer syndrome
NM_005708.4(GPC6):c.700C>T (p.Arg234Ter)	GPC6	13	93830534	Omodysplasia 1
NM_177987.2(TUBB8):c.1249G>A (p.Asp417Asn)	TUBB8	10	47143	Oocyte maturation defect 2
NM_177987.2(TUBB8):c.5G>A (p.Arg2Lys)	TUBB8	10	49234	Oocyte maturation defect 2
NM_177987.2(TUBB8):c.900G>A (p.Met300Ile)	TUBB8	10	47492	Oocyte maturation defect 2
NM_177987.2(TUBB8):c.785G>A (p.Arg262Gln)	TUBB8	10	47607	Oocyte maturation defect 2
NM_177987.2(TUBB8):c.527C>T (p.Ser176Leu)	TUBB8	10	47865	Oocyte maturation defect 2
NM_177987.2(TUBB8):c.713C>T (p.Thr238Met)	TUBB8	10	47679	Oocyte maturation defect 2
m.5703G>A	MT-TN	MT	5703	Ophthalmoplegia, isolated
NM_015330.4(SPECC1L):c.3247G>A (p.Gly1083Ser)	SPECC1L	22	24412690	Opitz G/BBB syndrome
NM_001567.3(INPPL1):c.1976C>T (p.Pro659Leu)	INPPL1	11	72233099	Opsismodysplasia
NM_001567.3(INPPL1):c.2415+1G>A	INPPL1	11	72234616	Opsismodysplasia
NM_001567.3(INPPL1):c.1975C>T (p.Pro659Ser)	INPPL1	11	72233098	Opsismodysplasia
NM_001567.3(INPPL1):c.1201C>T (p.Arg401Trp)	INPPL1	11	72230799	Opsismodysplasia
NM_001567.3(INPPL1):c.2071C>T (p.Arg691Trp)	INPPL1	11	72233471	Opsismodysplasia
NM_032730.5(RTN4IP1):c.308G>A (p.Arg103His)	RTN4IP1	6	106622936	Optic atrophy 10 with or without ataxia, mental retardation, and seizures
NM_139312.2(YME1L1):c.616C>T (p.Arg206Trp)	YME1L1	10	27136371	OPTIC ATROPHY 11
NM_032273.3(TMEM126A):c.163C>T (p.Arg55Ter)	TMEM126A	11	85654139	Optic atrophy 7
NM_001098.2(ACO2):c.1981G>A (p.Gly661Arg)	ACO2	22	41527315	Optic atrophy 9
NM_025136.3(OPA3):c.277G>A (p.Gly93Ser)	OPA3	19	45553777	Optic atrophy and cataract, autosomal dominant
NM_003106.3(SOX2):c.571G>A (p.Ala191Thr)	SOX2	3	181712931	Optic nerve hypoplasia and abnormalities of the central nervous system
NM_000280.4(PAX6):c.613C>T (p.Gln205Ter)	PAX6	11	31794699	Optic nerve hypoplasia, bilateral;not provided

NM_003611.2(OFD1):c.1099C>T (p.Arg367Ter)	OFD1	X	13753411	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.1100G>A (p.Arg367Gln)	OFD1	X	13753412	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.111G>A (p.Lys37=)	OFD1	X	13735346	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.121C>T (p.Arg41Ter)	OFD1	X	13736487	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.1420C>T (p.Gln474Ter)	OFD1	X	13757668	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.221C>T (p.Ser74Phe)	OFD1	X	13736587	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.235G>A (p.Ala79Thr)	OFD1	X	13736601	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.247C>T (p.Gln83Ter)	OFD1	X	13736613	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.337C>T (p.Gln113Ter)	OFD1	X	13738870	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.412G>A (p.Gly138Ser)	OFD1	X	13739032	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.454C>T (p.Gln152Ter)	OFD1	X	13744456	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.628C>T (p.Gln210Ter)	OFD1	X	13746429	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.823C>T (p.Gln275Ter)	OFD1	X	13746948	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.518-1G>A	OFD1	X	13746318	Orai-facial-digital syndrome
NM_003611.2(OFD1):c.1612C>T (p.Gln538*)	OFD1	X	13758406	Orai-facial-digital syndrome
NM_000274.3(OAT):c.3G>A (p.Met1Ile)	OAT	10	124412169	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.1186C>T (p.Arg396Ter)	OAT	10	124398076	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.812G>A (p.Arg271Lys)	OAT	10	124403015	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.1250C>T (p.Pro417Leu)	OAT	10	124398012	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.955C>T (p.His319Tyr)	OAT	10	124401785	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.550G>A (p.Ala184Thr)	OAT	10	124405534	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.722C>T (p.Pro241Leu)	OAT	10	124403847	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.1058G>A (p.Gly353Asp)	OAT	10	124400941	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.1172G>A (p.Trp391Ter)	OAT	10	124398090	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.1276C>T (p.Arg426Ter)	OAT	10	124397986	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.533G>A (p.Trp178Ter)	OAT	10	124405551	Ornithine aminotransferase deficiency
NM_000274.3(OAT):c.824G>A (p.Trp275Ter)	OAT	10	124403003	Ornithine aminotransferase deficiency
NM_000531.5(OTC):c.422G>A (p.Arg141Gln)	OTC	X	38401310	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.421C>T (p.Arg141Ter)	OTC	X	38401309	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.77G>A (p.Arg26Gln)	OTC	X	38352773	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.829C>T (p.Arg277Trp)	OTC	X	38408987	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.674C>T (p.Pro225Leu)	OTC	X	38408752	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.259G>A (p.Glu87Lys)	OTC	X	38369838	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.484G>A (p.Gly162Arg)	OTC	X	38401372	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.236G>A (p.Gly79Glu)	OTC	X	38369815	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.386G>A (p.Arg129His)	OTC	X	38381429	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.118C>T (p.Arg40Cys)	OTC	X	38367331	Ornithine carbamoyltransferase deficiency;not provided
NM_000531.5(OTC):c.119G>A (p.Arg40His)	OTC	X	38367332	Ornithine carbamoyltransferase deficiency;not provided
NM_001202.5(BMP4):c.1037C>T (p.Ala346Val)	BMP4	14	53950222	Orofacial cleft 11
NM_001202.5(BMP4):c.860G>A (p.Arg287His)	BMP4	14	53950399	Orofacial cleft 11
NM_002448.3(MSX1):c.365G>A (p.Gly122Glu)	MSX1	4	4860264	Orofacial cleft 5
NM_015631.5(TCTN3):c.1327C>T (p.Gln443Ter)	TCTN3	10	95682776	Orofacial-digital syndrome IV
NM_015631.5(TCTN3):c.877C>T (p.Gln293Ter)	TCTN3	10	95686506	Orofacial-digital syndrome IV
NM_001031725.5(DDX59):c.1600G>A (p.Gly534Arg)	DDX59	1	200644514	Orofaciodigital syndrome 5
NM_023073.3(C5orf42):c.3380C>T (p.Ser1127Leu)	C5orf42	5	37201718	Orofaciodigital syndrome 6

NM_015910.5(WDPCP):c.160G>A (p.Asp54Asn)	WDPCP	2	63492856	Orstavik Lindemann Solberg syndrome
NM_000118.3(ENG):c.68-1G>A	ENG	9	127843246	Osler hemorrhagic telangiectasia syndrome
NM_000118.3(ENG):c.991G>A (p.Gly331Ser)	ENG	9	127824800	Osler hemorrhagic telangiectasia syndrome
NM_000118.3(ENG):c.-127C>T	ENG	9	127854482	Osler hemorrhagic telangiectasia syndrome
NM_000118.3(ENG):c.1586G>A (p.Arg529His)	ENG	9	127818220	Osler hemorrhagic telangiectasia syndrome;not provided
NM_000118.3(ENG):c.360+1G>A	ENG	9	127829686	Osler hemorrhagic telangiectasia syndrome;not provided
NM_001463.3(FRZB):c.598C>T (p.Arg200Trp)	FRZB	2	182838608	Osteoarthritis susceptibility 1
NM_001844.4(COL2A1):c.2155C>T (p.Arg719Cys)	COL2A1	12	47982886	Osteoarthritis with mild chondrodysplasia
NM_013227.3(ACAN):c.7249G>A (p.Val2417Met)	ACAN	15	88872941	Osteochondritis dissecans
NR_023343.1(RNU4ATAC):n.55G>A	RNU4ATAC	2	121530934	Osteodysplastic primordial dwarfism, type 1
NR_023343.1(RNU4ATAC):r.30G>A	RNU4ATAC	2	121530909	Osteodysplastic primordial dwarfism, type 1
NR_023343.1(RNU4ATAC):n.111G>A	RNU4ATAC	2	121530990	Osteodysplastic primordial dwarfism, type 1
NR_023343.1(RNU4ATAC):n.50G>A	RNU4ATAC	2	121530929	Osteodysplastic primordial dwarfism, type 1
NR_023343.1(RNU4ATAC):n.124G>A	RNU4ATAC	2	121531003	Osteodysplastic primordial dwarfism, type 1
NR_023343.1(RNU4ATAC):n.51G>A	RNU4ATAC	2	121530930	Osteodysplastic primordial dwarfism, type 1;Roifman syndrome
NM_000088.3(COL1A1):c.787G>A (p.Gly263Arg)	COL1A1	17	50197027	Osteogenesis imperfecta
NM_000088.3(COL1A1):c.1200+1G>A	COL1A1	17	50195433	Osteogenesis imperfecta
NM_000088.3(COL1A1):c.3076C>T (p.Arg1026Ter)	COL1A1	17	50188765	Osteogenesis imperfecta
NM_000089.3(COL1A2):c.838G>A (p.Gly280Ser)	COL1A2	7	94409367	Osteogenesis imperfecta
NM_021939.3(FKBP10):c.1207C>T (p.Arg403Ter)	FKBP10	17	41820412	Osteogenesis imperfecta type 12
NM_001025295.2(IFITM5):c.-14C>T	IFITM5	11	299504	Osteogenesis imperfecta type 5
NM_001025295.2(IFITM5):c.100C>T (p.Arg34Ter)	IFITM5	11	299391	Osteogenesis imperfecta type 5
NM_001025295.2(IFITM5):c.119C>T (p.Ser40Leu)	IFITM5	11	299372	Osteogenesis imperfecta type 5;not provided
NM_006371.4(CRTAP):c.826C>T (p.Gln276Ter)	CRTAP	3	33129971	Osteogenesis imperfecta type 7
NM_006371.4(CRTAP):c.3G>A (p.Met1Ile)	CRTAP	3	33114080	Osteogenesis imperfecta type 7
NM_001146289.1(P3H1):c.2073G>A (p.Ala691=)	P3H1	1	42747254	Osteogenesis imperfecta type 8
NM_001146289.1(P3H1):c.1102C>T (p.Arg368Ter)	P3H1	1	42755616	Osteogenesis imperfecta type 8;not provided
NM_000942.4(PPIB):c.451C>T (p.Gln151Ter)	PPIB	15	64156802	Osteogenesis imperfecta type 9;not provided
NM_000942.4(PPIB):c.313G>A (p.Gly105Arg)	PPIB	15	64160134	Osteogenesis imperfecta type 9;not provided
NM_000088.3(COL1A1):c.3235G>A (p.Gly1079Ser)	COL1A1	17	50188122	Osteogenesis imperfecta type I
NM_000088.3(COL1A1):c.3421C>T (p.Arg1141Ter)	COL1A1	17	50187486	Osteogenesis imperfecta type I
NM_000088.3(COL1A1):c.2089C>T (p.Arg697Ter)	COL1A1	17	50191826	Osteogenesis imperfecta type I
NM_000088.3(COL1A1):c.91C>T (p.Gln31Ter)	COL1A1	17	50201423	Osteogenesis imperfecta type I;Osteogenesis imperfecta with normal sclerae, dominant form;Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.1678G>A (p.Gly560Ser)	COL1A1	17	50194032	Osteogenesis imperfecta type I;Osteogenesis imperfecta, recessive perinatal lethal;not provided
NM_000089.3(COL1A2):c.2575G>A (p.Gly859Ser)	COL1A2	7	94424345	Osteogenesis imperfecta type III
NM_000089.3(COL1A2):c.2251G>A (p.Gly751Ser)	COL1A2	7	94420604	Osteogenesis imperfecta type III
NM_000089.3(COL1A2):c.3269G>A (p.Gly1090Asp)	COL1A2	7	94427628	Osteogenesis imperfecta type III
NM_000089.3(COL1A2):c.3295G>A (p.Gly1099Arg)	COL1A2	7	94427654	Osteogenesis imperfecta type III
NM_000088.3(COL1A1):c.3064G>A (p.Gly1022Ser)	COL1A1	17	50188777	Osteogenesis imperfecta type III
NM_000088.3(COL1A1):c.1588G>A (p.Gly530Ser)	COL1A1	17	50194375	Osteogenesis imperfecta type III
NM_000088.3(COL1A1):c.3118G>A (p.Gly1040Ser)	COL1A1	17	50188619	Osteogenesis imperfecta type III
NM_000088.3(COL1A1):c.2515G>A (p.Gly839Ser)	COL1A1	17	50190045	Osteogenesis imperfecta type III
NM_000088.3(COL1A1):c.761G>A (p.Gly254Glu)	COL1A1	17	50197053	Osteogenesis imperfecta type III
NM_000089.3(COL1A2):c.1550G>A (p.Gly517Asp)	COL1A2	7	94413129	Osteogenesis imperfecta type III

NM_000089.3(COL1A2):c.1971+1G>A	COL1A2	7	94417832	Osteogenesis imperfecta type III;Osteogenesis imperfecta with normal sclerae, dominant form
NM_000089.3(COL1A2):c.2099G>A (p.Gly700Asp)	COL1A2	7	94420252	Osteogenesis imperfecta type III;Osteogenesis imperfecta with normal sclerae, dominant form
NM_000088.3(COL1A1):c.3028G>A (p.Gly1010Ser)	COL1A1	17	50188920	Osteogenesis imperfecta with normal sclerae, dominant form
NM_000089.3(COL1A2):c.2720G>A (p.Gly907Asp)	COL1A2	7	94425163	Osteogenesis imperfecta, recessive perinatal lethal
NM_000089.3(COL1A2):c.1640G>A (p.Gly547Asp)	COL1A2	7	94413922	Osteogenesis imperfecta, recessive perinatal lethal
NM_000089.3(COL1A2):c.2593G>A (p.Gly865Ser)	COL1A2	7	94424363	Osteogenesis imperfecta, recessive perinatal lethal
NM_000089.3(COL1A2):c.2414G>A (p.Gly805Asp)	COL1A2	7	94422967	Osteogenesis imperfecta, recessive perinatal lethal
NM_000089.3(COL1A2):c.1739G>A (p.Gly580Asp)	COL1A2	7	94415245	Osteogenesis imperfecta, recessive perinatal lethal
NM_000089.3(COL1A2):c.1504G>A (p.Gly502Ser)	COL1A2	7	94413083	Osteogenesis imperfecta, recessive perinatal lethal
NM_000089.3(COL1A2):c.1262G>A (p.Gly421Asp)	COL1A2	7	94411066	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.824G>A (p.Gly275Asp)	COL1A1	17	50196651	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.2210G>A (p.Gly737Asp)	COL1A1	17	50191408	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.2552G>A (p.Gly851Asp)	COL1A1	17	50190008	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.2533G>A (p.Gly845Arg)	COL1A1	17	50190027	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.3073G>A (p.Gly1025Arg)	COL1A1	17	50188768	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.3182G>A (p.Gly1061Asp)	COL1A1	17	50188555	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.3271G>A (p.Gly1091Ser)	COL1A1	17	50187974	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.3559G>A (p.Gly1187Ser)	COL1A1	17	50186895	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.2156G>A (p.Gly719Asp)	COL1A1	17	50191462	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.3541G>A (p.Gly1181Ser)	COL1A1	17	50186913	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.1777G>A (p.Gly593Ser)	COL1A1	17	50193038	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.1598G>A (p.Gly533Asp)	COL1A1	17	50194365	Osteogenesis imperfecta, recessive perinatal lethal
NM_000088.3(COL1A1):c.4160C>T (p.Ala1387Val)	COL1A1	17	50185866	Osteogenesis imperfecta, recessive perinatal lethal
NM_002615.6(SERPINF1):c.1132C>T (p.Gln378Ter)	SERPINF1	17	1777321	Osteogenesis imperfecta, type VI;not provided
NM_001309443.1(SPARC):c.494G>A (p.Arg165His)	SPARC	5	151667555	Osteogenesis imperfecta, type xvii
NM_001309444.1(SPARC):c.787G>A (p.Glu263Lys)	SPARC	5	151664183	Osteogenesis imperfecta, type xvii
NM_000088.3(COL1A1):c.2161C>T (p.Gln721Ter)	COL1A1	17	50191457	Osteogenesis imperfecta;Osteogenesis imperfecta type I
NM_000088.3(COL1A1):c.994G>A (p.Gly332Arg)	COL1A1	17	50196163	Osteogenesis imperfecta;Osteogenesis imperfecta type III;not provided
NM_173842.2(IL1RN):c.160C>T (p.Gln54Ter)	IL1RN	2	113129619	Osteomyelitis, sterile multifocal, with periostitis and pustulosis;not provided
NM_152424.3(AMER1):c.1057C>T (p.Arg353Ter)	AMER1	X	64192230	Osteopathia striata with cranial sclerosis
NM_152424.3(AMER1):c.811C>T (p.Gln271Ter)	AMER1	X	64192476	Osteopathia striata with cranial sclerosis
NM_152424.3(AMER1):c.1072C>T (p.Arg358Ter)	AMER1	X	64192215	Osteopathia striata with cranial sclerosis;not provided
NM_002335.3(LRP5):c.758C>T (p.Thr253Ile)	LRP5	11	68363818	Osteoporosis autosomal dominant type 1
NM_002335.3(LRP5):c.724G>A (p.Ala242Thr)	LRP5	11	68363784	Osteoporosis autosomal dominant type 1;Van Buchem disease type 2;Worth disease;not provided
NM_001287.5(CLCN7):c.2299C>T (p.Arg767Trp)	CLCN7	16	1447038	Osteopetrosis autosomal dominant type 2;Osteopetrosis autosomal recessive 4
NM_006019.3(TCIRG1):c.1213G>A (p.Gly405Arg)	TCIRG1	11	68047480	Osteopetrosis autosomal recessive 1
NM_006019.3(TCIRG1):c.1674-1G>A	TCIRG1	11	68049080	Osteopetrosis autosomal recessive 1
NM_001287.5(CLCN7):c.1663C>T (p.Gln555Ter)	CLCN7	16	1449282	Osteopetrosis autosomal recessive 4
NM_001287.5(CLCN7):c.2285G>A (p.Arg762Gln)	CLCN7	16	1447052	Osteopetrosis autosomal recessive 4
NM_001287.5(CLCN7):c.643G>A (p.Gly215Arg)	CLCN7	16	1459139	Osteopetrosis autosomal recessive 4
NM_014798.2(PLEKHM1):c.296+1G>A	PLEKHM1	17	45477899	Osteopetrosis autosomal recessive 6
NM_000067.2(CA2):c.319C>T (p.His107Tyr)	CA2	8	85473779	Osteopetrosis with renal tubular acidosis
NM_000067.2(CA2):c.232+1G>A	CA2	8	85465470	Osteopetrosis with renal tubular acidosis
NM_001199835.1(SNX10):c.152G>A (p.Arg51Gln)	SNX10	7	26364575	Osteopetrosis, autosomal recessive 8
NM_001199835.1(SNX10):c.46C>T (p.Arg16Ter)	SNX10	7	26360996	Osteopetrosis, autosomal recessive 8

NM_002335.3(LRP5):c.29G>A (p.Trp10Ter)	LRP5	11	68312743	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.1282C>T (p.Arg428Ter)	LRP5	11	68386582	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.2557C>T (p.Gln853Ter)	LRP5	11	68413742	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.1708C>T (p.Arg570Trp)	LRP5	11	68403606	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.2202G>A (p.Trp734Ter)	LRP5	11	68410024	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.1655C>T (p.Thr552Met)	LRP5	11	68403553	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.1145C>T (p.Pro382Leu)	LRP5	11	68386445	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.731C>T (p.Thr244Met)	LRP5	11	68363791	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.4600C>T (p.Arg1534Ter)	LRP5	11	68448822	Osteoporosis with pseudoglioma
NM_002335.3(LRP5):c.1481G>A (p.Arg494Gln)	LRP5	11	68389949	Osteoporosis with pseudoglioma;not provided
NM_000503.5(EYA1):c.639+1G>A	EYA1	8	71299637	Otofaciocervical syndrome
NM_001110556.1(FLNA):c.760G>A (p.Glu254Lys)	FLNA	X	154367505	Oto-palato-digital syndrome, type I;Oto-palato-digital syndrome, type II;Periventricular nodular heterotopia 1;not provided
NM_001110556.1(FLNA):c.2761C>T (p.Arg921Ter)	FLNA	X	154362044	Oto-palato-digital syndrome, type I;Periventricular nodular heterotopia 1;not provided
NM_001110556.1(FLNA):c.4543C>T (p.Arg1515Ter)	FLNA	X	154358500	Oto-palato-digital syndrome, type I;Periventricular nodular heterotopia 1;not provided
NM_001110556.1(FLNA):c.7153C>T (p.Gln2385Ter)	FLNA	X	154350912	Oto-palato-digital syndrome, type I;Periventricular nodular heterotopia 1;not provided
NM_080680.2(COL11A2):c.1981G>A (p.Gly661Arg)	COL11A2	6	33177216	Otospondylomegaepiphyseal dysplasia
NM_000145.3(FSHR):c.566C>T (p.Ala189Val)	FSHR	2	48983125	Ovarian dysgenesis 1
NM_000145.3(FSHR):c.1717C>T (p.Arg573Cys)	FSHR	2	48963104	Ovarian dysgenesis 1
NM_000145.3(FSHR):c.1255G>A (p.Ala419Thr)	FSHR	2	48963566	Ovarian dysgenesis 1
NM_017696.2(MCM9):c.394C>T (p.Arg132Ter)	MCM9	6	118924038	Ovarian dysgenesis 4;Premature ovarian failure 1
NM_000145.3(FSHR):c.1346C>T (p.Thr449Ile)	FSHR	2	48963475	Ovarian hyperstimulation syndrome
NM_000145.3(FSHR):c.1699G>A (p.Asp567Asn)	FSHR	2	48963122	Ovarian hyperstimulation syndrome
NM_014239.3(EIF2B2):c.547C>T (p.Arg183Ter)	EIF2B2	14	75004850	Ovarioleukodystrophy
NM_014239.3(EIF2B2):c.512C>T (p.Ser171Phe)	EIF2B2	14	75004815	Ovarioleukodystrophy
NM_003907.2(EIF2B5):c.583C>T (p.Arg195Cys)	EIF2B5	3	184137974	Ovarioleukodystrophy
NM_005555.3(KRT6B):c.1414G>A (p.Glu472Lys)	KRT6B	12	52447788	Pachyonychia congenita 4;not provided
NM_000422.2(KRT17):c.304G>A (p.Val102Met)	KRT17	17	41624206	Pachyonychia congenita type 2;not provided
NM_000422.2(KRT17):c.281G>A (p.Arg94His)	KRT17	17	41624229	Pachyonychia congenita type 2;Steatocystoma multiplex;not provided
NM_000422.2(KRT17):c.280C>T (p.Arg94Cys)	KRT17	17	41624230	Pachyonychia congenita type 2;Steatocystoma multiplex;not provided
NM_002977.3(SCN9A):c.1642C>T (p.Arg548Ter)	SCN9A	2	166284785	Pain insensitivity
NM_002977.3(SCN9A):c.3774+5C>T	SCN9A	2	166306951	Pain insensitivity
NM_000168.5(GLI3):c.2149C>T (p.Gln717Ter)	GLI3	7	41967878	Pallister-Hall syndrome
NM_000168.5(GLI3):c.2146C>T (p.Gln716Ter)	GLI3	7	41967881	Pallister-Hall syndrome
NM_000168.5(GLI3):c.2431+1G>A	GLI3	7	41967595	Pallister-Hall syndrome
NM_000168.5(GLI3):c.2110C>T (p.Gln704Ter)	GLI3	7	41967917	Pallister-Hall syndrome
NM_000168.5(GLI3):c.3481C>T (p.Gln1161Ter)	GLI3	7	41965592	Pallister-Hall syndrome
NM_001136191.2(KANK2):c.2009C>T (p.Ala670Val)	KANK2	19	11174532	Palmoplantar keratoderma and woolly hair
NM_000226.3(KRT9):c.478C>T (p.Leu160Phe)	KRT9	17	41571515	Palmoplantar keratoderma, epidermolytic, with knuckle pads;not provided
NM_145068.3(TRPV3):c.2017C>T (p.Leu673Phe)	TRPV3	17	3518644	Palmoplantar keratoderma, mutilating, with periorificial keratotic plaques
NM_145068.3(TRPV3):c.1717G>A (p.Gly573Ser)	TRPV3	17	3524224	Palmoplantar keratoderma, mutilating, with periorificial keratotic plaques;not provided
NM_001040147.2(SERPINB7):c.455-1G>A	SERPINB7	18	63798603	Palmoplantar keratoderma, nagashima type
NM_003784.3(SERPINB7):c.796C>T (p.Arg266Ter)	SERPINB7	18	63804288	Palmoplantar keratoderma, nagashima type;not provided
NM_173086.4(KRT6C):c.1414G>A (p.Glu472Lys)	KRT6C	12	52469680	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse;not provided
NM_005557.3(KRT16):c.379C>T (p.Arg127Cys)	KRT16	17	41612310	Palmoplantar keratoderma, nonepidermolytic, focal;not provided

NM_005257.5(GATA6):c.1367G>A (p.Arg456His)	GATA6	18	22181517	Pancreatic agenesis and congenital heart disease
NM_005257.5(GATA6):c.1399G>A (p.Ala467Thr)	GATA6	18	22181549	Pancreatic agenesis and congenital heart disease
NM_005257.5(GATA6):c.1087C>T (p.Gln363Ter)	GATA6	18	22172231	Pancreatic agenesis and congenital heart disease
NM_000209.3(PDX1):c.532G>A (p.Glu178Lys)	PDX1	13	27924381	Pancreatic agenesis, congenital
NM_002770.3(PRSS2):c.571G>A (p.Gly191Arg)	PRSS2	7	142774035	Pancreatitis, chronic, protection against
NM_001814.5(CTSC):c.628C>T (p.Arg210Ter)	CTSC	11	88309176	Papillon-LefÃ¨vre syndrome
NM_001814.5(CTSC):c.890-1G>A	CTSC	11	88294509	Papillon-LefÃ¨vre syndrome
NM_001814.5(CTSC):c.856C>T (p.Gln286Ter)	CTSC	11	88296166	Papillon-LefÃ¨vre syndrome
NM_001814.5(CTSC):c.901G>A (p.Gly301Ser)	CTSC	11	88294497	Papillon-LefÃ¨vre syndrome
NM_000148.3(FUT1):c.826C>T (p.Gln276Ter)	FUT1	19	48750456	Para-Bombay phenotype
NM_000148.3(FUT1):c.349C>T (p.His117Tyr)	FUT1	19	48750933	Para-bombay phenotype
NM_003002.3(SDHD):c.106C>T (p.Gln36Ter)	SDHD	11	112087910	Paragangliomas 1
NM_003002.3(SDHD):c.64C>T (p.Arg22Ter)	SDHD	11	112087868	Paragangliomas 1;Pheochromocytoma
NM_003002.3(SDHD):c.315G>A (p.Trp105Ter)	SDHD	11	112094805	Paragangliomas 1;Pheochromocytoma
NM_003002.3(SDHD):c.325C>T (p.Gln109Ter)	SDHD	11	112094815	Paragangliomas 1;Pheochromocytoma
NM_003001.3(SDHC):c.3G>A (p.Met1Ile)	SDHC	1	161314408	Paragangliomas 3
NM_003000.2(SDHB):c.343C>T (p.Arg115Ter)	SDHB	1	17028680	Paragangliomas 4;Pheochromocytoma
NM_004168.3(SDHA):c.1765C>T (p.Arg589Trp)	SDHA	5	251439	Paragangliomas 5
NM_000334.4(SCN4A):c.4367G>A (p.Gly1456Glu)	SCN4A	17	63941915	Paramyotonia congenita of von Eulenburg
NM_000334.4(SCN4A):c.4428G>A (p.Met1476Ile)	SCN4A	17	63941854	Paramyotonia congenita of von Eulenburg
NM_000334.4(SCN4A):c.3938C>T (p.Thr1313Met)	SCN4A	17	63943825	Paramyotonia congenita of von Eulenburg;not provided
NM_000334.4(SCN4A):c.3877G>A (p.Val1293Ile)	SCN4A	17	63944708	Paramyotonia congenita of von Eulenburg;not provided
NM_000334.4(SCN4A):c.4765G>A (p.Val1589Met)	SCN4A	17	63941517	Paramyotonia congenita of von Eulenburg;Potassium aggravated myotonia
NM_000334.4(SCN4A):c.2411C>T (p.Ser804Phe)	SCN4A	17	63951866	Paramyotonia congenita/myotonia congenita;Potassium aggravated myotonia
NM_000244.3(MEN1):c.76G>A (p.Glu26Lys)	MEN1	11	64810034	Parathyroid adenoma, somatic
NM_024529.4(CDC73):c.128G>A (p.Trp43Ter)	CDC73	1	193122328	Parathyroid carcinoma
NM_002449.4(MSX2):c.515G>A (p.Arg172His)	MSX2	5	174729294	Parietal foramina 1
NM_021926.3(ALX4):c.418C>T (p.Gln140Ter)	ALX4	11	44309645	Parietal foramina 2
NM_021926.3(ALX4):c.736C>T (p.Gln246Ter)	ALX4	11	44275389	Parietal foramina 2
NM_021926.3(ALX4):c.653G>A (p.Arg218Gln)	ALX4	11	44275472	Parietal foramina 2
NM_000345.3(SNCA):c.157G>A (p.Ala53Thr)	SNCA	4	89828149	Parkinson disease 1
NM_000345.3(SNCA):c.152G>A (p.Gly51Asp)	SNCA	4	89828154	Parkinson disease 1
NM_003560.3(PLA2G6):c.2222G>A (p.Arg741Gln)	PLA2G6	22	38112558	Parkinson disease 14
NM_003560.3(PLA2G6):c.2239C>T (p.Arg747Trp)	PLA2G6	22	38112541	Parkinson disease 14
NM_003560.3(PLA2G6):c.1904G>A (p.Arg635Gln)	PLA2G6	22	38115657	Parkinson disease 14
NM_003560.3(PLA2G6):c.1354C>T (p.Gln452Ter)	PLA2G6	22	38126444	Parkinson disease 14
NM_012179.3(FBXO7):c.1492C>T (p.Arg498Ter)	FBXO7	22	32498453	Parkinson disease 15
NM_012179.3(FBXO7):c.65C>T (p.Thr22Met)	FBXO7	22	32475067	Parkinson disease 15
NM_018206.5(VPS35):c.1858G>A (p.Asp620Asn)	VPS35	16	46662452	Parkinson disease 17
NM_198241.2(EIF4G1):c.3614G>A (p.Arg1205His)	EIF4G1	3	184327401	Parkinson disease 18
NM_198241.2(EIF4G1):c.1505C>T (p.Ala502Val)	EIF4G1	3	184322089	Parkinson disease 18
NM_001256865.1(DNAJC6):c.2200C>T (p.Gln734Ter)	DNAJC6	1	65406052	Parkinson disease 19a, juvenile-onset
NM_001256865.1(DNAJC6):c.2326C>T (p.Gln776Ter)	DNAJC6	1	65408685	Parkinson disease 19a, juvenile-onset
NM_004562.2(PRKN):c.931C>T (p.Gln311Ter)	PRKN	6	161569357	Parkinson disease 2
NM_004562.2(PRKN):c.1358G>A (p.Trp453Ter)	PRKN	6	161350139	Parkinson disease 2

NM_004562.2(PRKN):c.635G>A (p.Cys212Tyr)	PRKN	6	161973401	Parkinson disease 2
NM_203446.2(SYNJ1):c.773G>A (p.Arg258Gln)	SYNJ1	21	32695106	Parkinson disease 20, early-onset
NM_001320327.1(CHCHD2):c.182C>T (p.Thr61Ile)	CHCHD2	7	56104344	Parkinson disease 22, autosomal dominant
NM_001320327.1(CHCHD2):c.434G>A (p.Arg145Gln)	CHCHD2	7	56102878	Parkinson disease 22, autosomal dominant
NM_001320327.1(CHCHD2):c.300+5G>A	CHCHD2	7	56104221	Parkinson disease 22, autosomal dominant
NM_032409.2(PINK1):c.926G>A (p.Gly309Asp)	PINK1	1	20644639	Parkinson disease 6, autosomal recessive early-onset
NM_032409.2(PINK1):c.1311G>A (p.Trp437Ter)	PINK1	1	20649054	Parkinson disease 6, autosomal recessive early-onset
NM_032409.2(PINK1):c.736C>T (p.Arg246Ter)	PINK1	1	20639952	Parkinson disease 6, autosomal recessive early-onset
NM_032409.2(PINK1):c.836G>A (p.Arg279His)	PINK1	1	20644549	Parkinson disease 6, autosomal recessive early-onset
NM_032409.2(PINK1):c.938C>T (p.Thr313Met)	PINK1	1	20644651	Parkinson disease 6, autosomal recessive early-onset
NM_032409.2(PINK1):c.1366C>T (p.Gln456Ter)	PINK1	1	20649109	Parkinson disease 6, autosomal recessive early-onset
NM_007262.4(PARK7):c.78G>A (p.Met26Ile)	PARK7	1	7962863	Parkinson disease 7
NM_198578.3(LRRK2):c.4321C>T (p.Arg1441Cys)	LRRK2	12	40310434	Parkinson disease 8, autosomal dominant
NM_198578.3(LRRK2):c.4322G>A (p.Arg1441His)	LRRK2	12	40310435	Parkinson disease 8, autosomal dominant
NM_198578.3(LRRK2):c.1256C>T (p.Ala419Val)	LRRK2	12	40252984	Parkinson disease 8, autosomal dominant
NM_198578.3(LRRK2):c.6055G>A (p.Gly2019Ser)	LRRK2	12	40340400	Parkinson disease 8, autosomal dominant;not provided
NM_022089.3(ATP13A2):c.1306+5G>A	ATP13A2	1	16996381	Parkinson disease 9
NM_022089.3(ATP13A2):c.2629G>A (p.Gly877Arg)	ATP13A2	1	16988455	Parkinson disease 9
NM_022089.3(ATP13A2):c.490C>T (p.Arg164Trp)	ATP13A2	1	17004399	Parkinson disease 9
NM_001005741.2(GBA):c.1444G>A (p.Asp482Asn)	GBA	1	155235256	Parkinson disease, late-onset
m.15950G>A	MT-TT	MT	15950	Parkinson disease, mitochondrial
NM_001199107.1(TBC1D24):c.1078C>T (p.Arg360Cys)	TBC1D24	16	2498332	Parkinsonism
NM_005765.2(ATP6AP2):c.345C>T (p.Ser115=)	ATP6AP2	X	40597293	Parkinsonism with spasticity, X-linked
NM_171998.3(RAB39B):c.574G>A (p.Gly192Arg)	RAB39B	X	155260871	Parkinsonism, early onset with mental retardation;X-linked dominant Parkinson's disease
NM_015488.4(PNKD):c.20C>T (p.Ala7Val)	PNKD	2	218270555	Paroxysmal choreoathetosis
NM_002977.3(SCN9A):c.2986C>T (p.Arg996Cys)	SCN9A	2	166272731	Paroxysmal extreme pain disorder
NM_002977.3(SCN9A):c.4391C>T (p.Thr1464Ile)	SCN9A	2	166204439	Paroxysmal extreme pain disorder
NM_001167623.1(CACNA1C):c.1204G>A (p.Gly402Ser)	CACNA1C	12	2504526	Paroxysmal familial ventricular fibrillation 1;not provided
NM_002641.3(PIGA):c.163C>T (p.Gln55Ter)	PIGA	X	15331768	Paroxysmal nocturnal hemoglobinuria 1
NM_000022.3(ADA):c.446G>A (p.Arg149Gln)	ADA	20	44625601	Partial adenosine deaminase deficiency;Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.821C>T (p.Pro274Leu)	ADA	20	44622612	Partial adenosine deaminase deficiency;Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.631C>T (p.Arg211Cys)	ADA	20	44623054	Partial adenosine deaminase deficiency;Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.643G>A (p.Ala215Thr)	ADA	20	44623042	Partial adenosine deaminase deficiency;Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.698C>T (p.Thr233Ile)	ADA	20	44622911	Partial adenosine deaminase deficiency;Severe combined immunodeficiency due to ADA deficiency
NM_000222.2(KIT):c.1990G>A (p.Gly664Arg)	KIT	4	54728121	Partial albinism
NM_000222.2(KIT):c.1747G>A (p.Glu583Lys)	KIT	4	54727515	Partial albinism
NM_000222.2(KIT):c.1879+1G>A	KIT	4	54727928	Partial albinism
NM_000194.2(HPRT1):c.329C>T (p.Ser110Leu)	HPRT1	X	134486475	Partial hypoxanthine-guanine phosphoribosyltransferase deficiency
NM_000194.2(HPRT1):c.503C>T (p.Thr168Ile)	HPRT1	X	134498407	Partial hypoxanthine-guanine phosphoribosyltransferase deficiency
NM_000194.2(HPRT1):c.46G>A (p.Gly16Ser)	HPRT1	X	134473377	Partial hypoxanthine-guanine phosphoribosyltransferase deficiency
NM_000194.2(HPRT1):c.172G>A (p.Gly58Arg)	HPRT1	X	134475218	Partial hypoxanthine-guanine phosphoribosyltransferase deficiency
NM_000194.2(HPRT1):c.193C>T (p.Leu65Phe)	HPRT1	X	134475239	Partial hypoxanthine-guanine phosphoribosyltransferase deficiency
NM_001136239.3(PRDM6):c.1646G>A (p.Arg549Gln)	PRDM6	5	123180296	Patent ductus arteriosus 3
NM_000322.4(PRPH2):c.500G>A (p.Gly167Asp)	PRPH2	6	42721835	Patterned dystrophy of retinal pigment epithelium;not provided
NM_005554.3(KRT6A):c.1414G>A (p.Glu472Lys)	KRT6A	12	52488338	PC-K6a;not provided

NM_001127895.1(CHST8):c.229C>T (p.Arg77Trp)	CHST8	19	33772017	Peeling skin syndrome 3
NM_198833.1(SERPINB8):c.850C>T (p.Arg284Ter)	SERPINB8	18	63987003	PEELING SKIN SYNDROME 5
NM_194442.2(LBR):c.356C>T (p.Pro119Leu)	LBR	1	225422087	Pelger-HuÃ¢t anomaly
NM_001128834.2(PLP1):c.44C>T (p.Pro15Leu)	PLP1	X	103785621	Pelizaeus-Merzbacher disease
NM_001128834.2(PLP1):c.467C>T (p.Thr156Ile)	PLP1	X	103787811	Pelizaeus-Merzbacher disease
NM_001128834.2(PLP1):c.220G>A (p.Gly74Arg)	PLP1	X	103786493	Pelizaeus-Merzbacher disease
NM_001128834.2(PLP1):c.128C>T (p.Thr43Ile)	PLP1	X	103785705	Pelizaeus-Merzbacher disease
NM_001128834.2(PLP1):c.453G>A (p.Lys151=)	PLP1	X	103786726	Pelizaeus-Merzbacher disease
NM_001128834.2(PLP1):c.434G>A (p.Trp145Ter)	PLP1	X	103786707	Pelizaeus-Merzbacher disease, atypical
NM_001128834.2(PLP1):c.725C>T (p.Ala242Val)	PLP1	X	103789361	Pelizaeus-Merzbacher disease, connatal
NM_001128834.2(PLP1):c.3G>A (p.Met1Ile)	PLP1	X	103776998	Pelizaeus-Merzbacher disease, mild
NM_001128834.2(PLP1):c.646C>T (p.Pro216Ser)	PLP1	X	103788460	Pelizaeus-Merzbacher disease;not provided
NM_005055.4(RAPSN):c.566C>T (p.Ala189Val)	RAPSN	11	47442780	Pena-Shokeir syndrome type I
NM_000441.1(SLC26A4):c.249G>A (p.Trp83Ter)	SLC26A4	7	107663380	Pendred's syndrome
NM_000441.1(SLC26A4):c.281C>T (p.Thr94Ile)	SLC26A4	7	107663412	Pendred's syndrome
NM_000441.1(SLC26A4):c.416-1G>A	SLC26A4	7	107674163	Pendred's syndrome
NM_001814.5(CTSC):c.815G>A (p.Arg272His)	CTSC	11	88296207	Periodontitis, aggressive, 1
NM_006941.3(SOX10):c.748C>T (p.Gln250Ter)	SOX10	22	37974148	Peripheral demyelinating neuropathy, central dysmyelination, Waardenburg syndrome, and Hirschsprung disease
NM_001110556.1(FLNA):c.544C>T (p.Gln182Ter)	FLNA	X	154367920	Periventricular nodular heterotopia 1
NM_001110556.1(FLNA):c.373+1G>A	FLNA	X	154370872	Periventricular nodular heterotopia 1
NM_001110556.1(FLNA):c.1966C>T (p.Leu656Phe)	FLNA	X	154364582	Periventricular nodular heterotopia 1
NM_001110556.1(FLNA):c.383C>T (p.Ala128Val)	FLNA	X	154368081	Periventricular nodular heterotopia 1
NM_001110556.1(FLNA):c.7896G>A (p.Trp2632Ter)	FLNA	X	154348897	Periventricular nodular heterotopia 1
NM_001110556.1(FLNA):c.4450C>T (p.Gln1484Ter)	FLNA	X	154359008	Periventricular nodular heterotopia 1
NM_001456.3(FLNA):c.6700C>T (p.Arg2234Ter)	FLNA	X	154352226	Periventricular nodular heterotopia 1
NM_001144967.2(NEDD4L):c.2690G>A (p.Arg897Gln)	NEDD4L	18	58390680	PERIVENTRICULAR NODULAR HETEROTOPIA 7;Periventricular nodular heterotopia with syndactyly, cleft palate and developmental delay
NM_001144967.2(NEDD4L):c.2677G>A (p.Glu893Lys)	NEDD4L	18	58390667	PERIVENTRICULAR NODULAR HETEROTOPIA 7;Periventricular nodular heterotopia with syndactyly, cleft palate and developmental delay;not provided
NM_000352.4(ABCC8):c.1144G>A (p.Glu382Lys)	ABCC8	11	17453151	Permanent neonatal diabetes mellitus
NM_001287174.1(ABCC8):c.134C>T (p.Pro45Leu)	ABCC8	11	17476643	Permanent neonatal diabetes mellitus
NM_000207.2(INS):c.287G>A (p.Cys96Tyr)	INS	11	2159898	Permanent neonatal diabetes mellitus
NM_000162.3(GCK):c.1133C>T (p.Ala378Val)	GCK	7	44145617	Permanent neonatal diabetes mellitus
NM_000162.3(GCK):c.790G>A (p.Gly264Ser)	GCK	7	44147723	Permanent neonatal diabetes mellitus
NM_000207.2(INS):c.265C>T (p.Arg89Cys)	INS	11	2159920	Permanent neonatal diabetes mellitus
NM_000525.3(KCNJ11):c.1001G>A (p.Gly334Asp)	KCNJ11	11	17387091	Permanent neonatal diabetes mellitus
NM_000525.3(KCNJ11):c.497G>A (p.Cys166Tyr)	KCNJ11	11	17387595	Permanent neonatal diabetes mellitus
NM_000207.2(INS):c.3G>A (p.Met1Ile)	INS	11	2160969	Permanent neonatal diabetes mellitus
NM_000207.2(INS):c.187+241G>A	INS	11	2160544	Permanent neonatal diabetes mellitus
NM_000207.2(INS):c.188-31G>A	INS	11	2160028	Permanent neonatal diabetes mellitus;not provided
NM_000286.2(PEX12):c.949C>T (p.Leu317Phe)	PEX12	17	35575913	Peroxisomal biogenesis disorder 3b
NM_000286.2(PEX12):c.538C>T (p.Arg180Ter)	PEX12	17	35577180	Peroxisomal biogenesis disorder 3b;Peroxisome biogenesis disorder 3A
NM_032228.5(FAR1):c.787C>T (p.Arg263Ter)	FAR1	11	13711946	Peroxisomal fatty acyl-coa reductase 1 disorder
NM_003630.2(PEX3):c.898C>T (p.Arg300Ter)	PEX3	6	143479155	PEROXISOME BIOGENESIS DISORDER 10B

NM_003630.2(PEX3):c.991G>A (p.Gly331Arg)	PEX3	6	143485201	PEROXISOME BIOGENESIS DISORDER 10B
NM_002618.3(PEX13):c.702G>A (p.Trp234Ter)	PEX13	2	61032028	Peroxisome biogenesis disorder 11A
NM_004565.2(PEX14):c.553C>T (p.Gln185Ter)	PEX14	1	10624405	Peroxisome biogenesis disorder 13A
NM_003846.2(PEX11B):c.64C>T (p.Gln22Ter)	PEX11B	1	145917809	Peroxisome biogenesis disorder 14B
NM_000466.2(PEX1):c.547C>T (p.Arg183Ter)	PEX1	7	92517968	Peroxisome biogenesis disorder 1B;Zellweger syndrome
NM_001131025.1(PEX5):c.1279C>T (p.Arg427Ter)	PEX5	12	7208554	Peroxisome biogenesis disorder 2a (zellweger);not provided
NM_000287.3(PEX6):c.2440C>T (p.Arg814Ter)	PEX6	6	42965712	Peroxisome biogenesis disorder 4a (zellweger)
NM_001079867.1(PEX2):c.163G>A (p.Glu55Lys)	PEX2	8	76984016	Peroxisome biogenesis disorder 5B
NM_001079867.1(PEX2):c.669G>A (p.Trp223Ter)	PEX2	8	76983510	Peroxisome biogenesis disorder 5B
NM_000318.2(PEX2):c.355C>T (p.Arg119Ter)	PEX2	8	76983824	Peroxisome biogenesis disorder 5B;Peroxisome biogenesis disorder 5a (zellweger)
NM_002617.3(PEX10):c.600+1G>A	PEX10	1	2408451	Peroxisome biogenesis disorder 6A
NM_153818.1(PEX10):c.373C>T (p.Arg125Ter)	PEX10	1	2408679	Peroxisome biogenesis disorder 6B
NM_153818.1(PEX10):c.992G>A (p.Arg331Gln)	PEX10	1	2405815	Peroxisome biogenesis disorder 6B
NM_153818.1(PEX10):c.790C>T (p.Arg264Ter)	PEX10	1	2406766	Peroxisome biogenesis disorder 6B
NM_017929.5(PEX26):c.265G>A (p.Gly89Arg)	PEX26	22	18079908	Peroxisome biogenesis disorder 7A
NM_017929.5(PEX26):c.296G>A (p.Trp99Ter)	PEX26	22	18079939	Peroxisome biogenesis disorder 7A
NM_017929.5(PEX26):c.292C>T (p.Arg98Trp)	PEX26	22	18079935	Peroxisome biogenesis disorder 7A;Peroxisome biogenesis disorder 7B
NM_004813.2(PEX16):c.526C>T (p.Arg176Ter)	PEX16	11	45914619	Peroxisome biogenesis disorder 8A;not provided
NM_000288.3(PEX7):c.653C>T (p.Ala218Val)	PEX7	6	136869909	Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata type 1
NM_000288.3(PEX7):c.649G>A (p.Gly217Arg)	PEX7	6	136869905	Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata type 1
NM_015340.3(LARS2):c.1886C>T (p.Thr629Met)	LARS2	3	45516118	Perrault syndrome 4
NM_015340.3(LARS2):c.899C>T (p.Thr300Met)	LARS2	3	45476508	Perrault syndrome 4
NM_015340.3(LARS2):c.1912G>A (p.Glu638Lys)	LARS2	3	45516144	Perrault syndrome 4
NM_021830.4(TWNK):c.1519G>A (p.Val507Ile)	TWNK	10	100990470	Perrault syndrome 5
NM_004082.4(DCTN1):c.211G>A (p.Gly71Arg)	DCTN1	2	74378068	Perry syndrome
NM_004082.4(DCTN1):c.212G>A (p.Gly71Glu)	DCTN1	2	74378067	Perry syndrome
NM_004082.4(DCTN1):c.200G>A (p.Gly67Asp)	DCTN1	2	74378079	Perry syndrome
NM_001287174.1(ABCC8):c.4310G>A (p.Arg1437Gln)	ABCC8	11	17395610	Persistent hyperinsulinemic hypoglycemia of infancy
NM_001287174.1(ABCC8):c.4261C>T (p.Arg1421Cys)	ABCC8	11	17395659	Persistent hyperinsulinemic hypoglycemia of infancy
NM_000352.4(ABCC8):c.4477C>T (p.Arg1493Trp)	ABCC8	11	17394334	Persistent hyperinsulinemic hypoglycemia of infancy
NM_000352.4(ABCC8):c.4119+1G>A	ABCC8	11	17396915	Persistent hyperinsulinemic hypoglycemia of infancy
NM_000352.4(ABCC8):c.2117-1G>A	ABCC8	11	17427155	Persistent hyperinsulinemic hypoglycemia of infancy
NM_000352.4(ABCC8):c.331G>A (p.Gly111Arg)	ABCC8	11	17470182	Persistent hyperinsulinemic hypoglycemia of infancy
NM_000352.4(ABCC8):c.3748C>T (p.Arg1250Ter)	ABCC8	11	17398344	Persistent hyperinsulinemic hypoglycemia of infancy
NM_001287174.1(ABCC8):c.3992-9G>A	ABCC8	11	17397055	Persistent hyperinsulinemic hypoglycemia of infancy;not provided
NM_001287174.1(ABCC8):c.4519G>A (p.Glu1507Lys)	ABCC8	11	17394295	Persistent hyperinsulinemic hypoglycemia of infancy;not provided
NM_001287174.1(ABCC8):c.2800C>T (p.Arg934Ter)	ABCC8	11	17408415	Persistent hyperinsulinemic hypoglycemia of infancy;not provided
NM_001287174.1(ABCC8):c.2509C>T (p.Arg837Ter)	ABCC8	11	17412716	Persistent hyperinsulinemic hypoglycemia of infancy;not provided
NM_000479.4(AMH):c.571C>T (p.Arg191Ter)	AMH	19	2250667	Persistent mullerian duct syndrome, type I
NM_020547.3(AMHR2):c.1217G>A (p.Arg406Gln)	AMHR2	12	53429907	Persistent mullerian duct syndrome, type II
NM_194318.3(B3GLCT):c.347+5G>A	B3GLCT	13	31247104	Peters plus syndrome
NM_194318.3(B3GLCT):c.1178G>A (p.Gly393Glu)	B3GLCT	13	31317679	Peters plus syndrome
NM_194318.3(B3GLCT):c.1065-1G>A	B3GLCT	13	31317565	Peters plus syndrome
NM_194318.3(B3GLCT):c.660+1G>A	B3GLCT	13	31269278	Peters plus syndrome;not provided
NM_003916.4(AP1S2):c.105C>T (p.Gln36Ter)	AP1S2	X	15852419	Pettigrew syndrome

NM_003916.4(AP1S2):c.154C>T (p.Arg52Ter)	AP1S2	X	15852371	Pettigrew syndrome	
NM_003916.4(AP1S2):c.288+5G>A	AP1S2	X	15845898	Pettigrew syndrome	
NM_000455.4(STK11):c.465-1G>A	STK11	19	1220372	Peutz-Jeghers syndrome	
NM_000141.4(FGFR2):c.940-1G>A	FGFR2	10	121517464	Pfeiffer syndrome	
NM_000277.1(PAH):c.1315+1G>A	PAH	12	102840399	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1222C>T (p.Arg408Trp)	PAH	12	102840493	Phenylketonuria;not provided	
NM_000277.1(PAH):c.977G>A (p.Trp326Ter)	PAH	12	102844424	Phenylketonuria;not provided	
NM_000277.1(PAH):c.838G>A (p.Glu280Lys)	PAH	12	102852819	Phenylketonuria;not provided	
NM_000277.1(PAH):c.331C>T (p.Arg111Ter)	PAH	12	102894756	Phenylketonuria;not provided	
NM_000277.1(PAH):c.782G>A (p.Arg261Gln)	PAH	12	102852875	Phenylketonuria;not provided	
NM_000277.1(PAH):c.754C>T (p.Arg252Trp)	PAH	12	102852903	Phenylketonuria;not provided	
NM_000277.1(PAH):c.473G>A (p.Arg158Gln)	PAH	12	102866632	Phenylketonuria;not provided	
NM_000277.1(PAH):c.727C>T (p.Arg243Ter)	PAH	12	102852930	Phenylketonuria;not provided	
NM_000277.1(PAH):c.842C>T (p.Pro281Leu)	PAH	12	102852815	Phenylketonuria;not provided	
NM_000277.1(PAH):c.728G>A (p.Arg243Gln)	PAH	12	102852929	Phenylketonuria;not provided	
NM_000277.1(PAH):c.442-1G>A	PAH	12	102866664	Phenylketonuria;not provided	
NM_000277.1(PAH):c.818C>T (p.Ser273Phe)	PAH	12	102852839	Phenylketonuria;not provided	
NM_000277.1(PAH):c.842+1G>A	PAH	12	102852814	Phenylketonuria;not provided	
NM_000277.1(PAH):c.776C>T (p.Ala259Val)	PAH	12	102852881	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1066-11G>A	PAH	12	102843790	Phenylketonuria;not provided	
NM_000277.1(PAH):c.781C>T (p.Arg261Ter)	PAH	12	102852876	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1223G>A (p.Arg408Gln)	PAH	12	102840492	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1162G>A (p.Val388Met)	PAH	12	102843683	Phenylketonuria;not provided	
NM_000277.1(PAH):c.731C>T (p.Pro244Leu)	PAH	12	102852926	Phenylketonuria;not provided	
NM_000277.1(PAH):c.3G>A (p.Met1Ile)	PAH	12	102917128	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1066-3C>T	PAH	12	102843782	Phenylketonuria;not provided	
NM_000277.1(PAH):c.136G>A (p.Gly46Ser)	PAH	12	102912823	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1220C>T (p.Pro407Leu)	PAH	12	102840495	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1200-1G>A	PAH	12	102840516	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1208C>T (p.Ala403Val)	PAH	12	102840507	Phenylketonuria;not provided	
NM_000277.1(PAH):c.890G>A (p.Arg297His)	PAH	12	102851709	Phenylketonuria;not provided	
NM_000277.1(PAH):c.912+1G>A	PAH	12	102851686	Phenylketonuria;not provided	
NM_000277.1(PAH):c.926C>T (p.Ala309Val)	PAH	12	102846938	Phenylketonuria;not provided	
NM_000277.1(PAH):c.1065+1G>A	PAH	12	102844335	Phenylketonuria;not provided	
NM_000277.1(PAH):c.441+1G>A	PAH	12	102877461	Phenylketonuria;not provided	
NM_000277.1(PAH):c.472C>T (p.Arg158Trp)	PAH	12	102866633	Phenylketonuria;not provided	
NM_000277.1(PAH):c.526C>T (p.Arg176Ter)	PAH	12	102855316	Phenylketonuria;not provided	
NM_000277.1(PAH):c.688G>A (p.Val230Ile)	PAH	12	102855154	Phenylketonuria;not provided	
NM_000277.1(PAH):c.721C>T (p.Arg241Cys)	PAH	12	102852936	Phenylketonuria;not provided	
NM_000277.1(PAH):c.745C>T (p.Leu249Phe)	PAH	12	102852912	Phenylketonuria;not provided	
NM_000277.1(PAH):c.755G>A (p.Arg252Gln)	PAH	12	102852902	Phenylketonuria;not provided	
NM_000277.1(PAH):c.809G>A (p.Arg270Lys)	PAH	12	102852848	Phenylketonuria;not provided	
NM_003002.3(SDHD):c.14G>A (p.Trp5Ter)	SDHD	11	112086921	Pheochromocytoma	
NM_003000.2(SDHB):c.302G>A (p.Cys101Tyr)	SDHB	1	17028721	Pheochromocytoma	
NM_003000.2(SDHB):c.299C>T (p.Ser100Phe)	SDHB	1	17028724	Pheochromocytoma	

NM_002382.4(MAX):c.97C>T (p.Arg33Ter)	MAX	14	65093782	Pheochromocytoma, susceptibility to
NM_000551.3(VHL):c.277G>A (p.Gly93Ser)	VHL	3	10142124	Pheochromocytoma;Von Hippel-Lindau syndrome
NM_006623.3(PHGDH):c.1468G>A (p.Val490Met)	PHGDH	1	119743906	Phosphoglycerate dehydrogenase deficiency
NM_006623.3(PHGDH):c.1273G>A (p.Val425Met)	PHGDH	1	119742870	Phosphoglycerate dehydrogenase deficiency
NM_006623.3(PHGDH):c.403C>T (p.Arg135Trp)	PHGDH	1	119726897	Phosphoglycerate dehydrogenase deficiency
NM_006623.3(PHGDH):c.1129G>A (p.Gly377Ser)	PHGDH	1	119741817	Phosphoglycerate dehydrogenase deficiency
NM_006623.3(PHGDH):c.781G>A (p.Val261Met)	PHGDH	1	119735432	Phosphoglycerate dehydrogenase deficiency
NM_000291.3(PGK1):c.802G>A (p.Asp268Asn)	PGK1	X	78123240	Phosphoglycerate kinase 1 deficiency
NM_000291.3(PGK1):c.959G>A (p.Ser320Asn)	PGK1	X	78124896	Phosphoglycerate kinase 1 deficiency
NM_000291.3(PGK1):c.756+5G>A	PGK1	X	78122954	Phosphoglycerate kinase 1 deficiency
NM_153373.3(PHYKPL):c.718G>A (p.Gly240Arg)	PHYKPL	5	178222564	Phosphohydroxylysinuria
NM_024420.2(PLA2G4A):c.1454G>A (p.Arg485His)	PLA2G4A	1	186956219	Phospholipase a2, group IV a, deficiency of
NM_002764.3(PRPS1):c.569C>T (p.Ala190Val)	PRPS1	X	107645215	Phosphoribosylpyrophosphate synthetase superactivity
NM_000288.3(PEX7):c.-45C>T	PEX7	6	136822621	Phytanic acid storage disease
NM_016835.4(MAPT):c.2116G>A (p.Gly706Arg)	MAPT	17	46024010	Pick's disease
NM_016835.4(MAPT):c.1910C>T (p.Ser637Phe)	MAPT	17	46014286	Pick's disease;not provided
NM_002292.3(LAMB2):c.736C>T (p.Arg246Trp)	LAMB2	3	49131129	Pierson syndrome
NM_002292.3(LAMB2):c.2890C>T (p.Arg964Ter)	LAMB2	3	49124920	Pierson syndrome
NM_153638.3(PANK2):c.790C>T (p.Arg264Trp)	PANK2	20	3908087	Pigmentary pallidal degeneration
NM_153638.3(PANK2):c.856C>T (p.Arg286Cys)	PANK2	20	3908153	Pigmentary pallidal degeneration
NM_153638.3(PANK2):c.1412G>A (p.Ser471Asn)	PANK2	20	3912634	Pigmentary pallidal degeneration
NM_002905.3(RDH5):c.285G>A (p.Trp95Ter)	RDH5	12	55721469	Pigmentary retinal dystrophy
NM_000326.4(RLBP1):c.452G>A (p.Arg151Gln)	RLBP1	15	89215133	Pigmentary retinal dystrophy;Retinitis punctata albescens
m.12183G>A	MT-TH	MT	12183	Pigmentary retinopathy and sensorineural deafness
NM_002734.4(PRKAR1A):c.-7+1G>A	PRKAR1A	17	68512549	Pigmented nodular adrenocortical disease, primary, 1
NM_004328.4(BCS1L):c.548G>A (p.Arg183His)	BCS1L	2	218661846	Pili torti-deafness syndrome
NM_000208.3(INSR):c.3946>A (p.Gly132Ser)	INSR	19	7267603	Pineal hyperplasia AND diabetes mellitus syndrome
NM_177438.2(DICER1):c.4050+1G>A	DICER1	14	95103345	Pineoblastoma
NM_001083962.1(TCF4):c.1086G>A (p.Trp362Ter)	TCF4	18	55257375	Pitt-Hopkins syndrome
NM_001083962.1(TCF4):c.1146+1G>A	TCF4	18	55257314	Pitt-Hopkins syndrome
NM_001083962.1(TCF4):c.655+1G>A	TCF4	18	55279550	Pitt-Hopkins syndrome
NM_001083962.1(TCF4):c.748C>T (p.Gln250Ter)	TCF4	18	55275660	Pitt-Hopkins syndrome
NM_001083962.1(TCF4):c.1438C>T (p.Gln480Ter)	TCF4	18	55234596	Pitt-Hopkins syndrome
NM_001083962.1(TCF4):c.968C>T (p.Ala323Val)	TCF4	18	55261488	Pitt-Hopkins syndrome
NM_001083962.1(TCF4):c.1411C>T (p.Gln471Ter)	TCF4	18	55234623	Pitt-Hopkins syndrome
NM_001083962.1(TCF4):c.1739G>A (p.Arg580Gln)	TCF4	18	55228987	Pitt-Hopkins syndrome;not provided
NM_001083962.1(TCF4):c.1153C>T (p.Arg385Ter)	TCF4	18	55254694	Pitt-Hopkins syndrome;not provided
NM_001083962.1(TCF4):c.1733G>A (p.Arg578His)	TCF4	18	55228993	Pitt-Hopkins syndrome;not provided
NM_001083962.1(TCF4):c.469C>T (p.Arg157Ter)	TCF4	18	55350904	Pitt-Hopkins syndrome;not provided
NM_001083962.1(TCF4):c.1738C>T (p.Arg580Trp)	TCF4	18	55228988	Pitt-Hopkins syndrome;Severe intellectual deficiency;not provided
NM_003977.3(AIP):c.40C>T (p.Gln14Ter)	AIP	11	67483198	Pituitary adenoma predisposition;Prolactinoma, familial;Somatotroph adenoma
NM_001122757.2(POU1F1):c.592C>T (p.Arg198Ter)	POU1F1	3	87262161	Pituitary hormone deficiency, combined 1
NM_001122757.2(POU1F1):c.889C>T (p.Arg297Trp)	POU1F1	3	87259959	Pituitary hormone deficiency, combined 1
NM_001122757.2(POU1F1):c.71C>T (p.Pro24Leu)	POU1F1	3	87276392	Pituitary hormone deficiency, combined 1
NM_001122757.2(POU1F1):c.506G>A (p.Arg169Gln)	POU1F1	3	87264299	Pituitary hormone deficiency, combined 1

NM_001122757.2(POU1F1):c.793C>T (p.Pro265Ser)	POU1F1	3	87260055	Pituitary hormone deficiency, combined 1	
NM_001122757.2(POU1F1):c.766G>A (p.Glu256Lys)	POU1F1	3	87260082	Pituitary hormone deficiency, combined 1	
NM_001122757.2(POU1F1):c.593G>A (p.Arg198Gln)	POU1F1	3	87262160	Pituitary hormone deficiency, combined 1	
NM_006261.4(PROP1):c.358C>T (p.Arg120Cys)	PROP1	5	177993032	Pituitary hormone deficiency, combined 2	
NM_006261.4(PROP1):c.218G>A (p.Arg73His)	PROP1	5	177994230	Pituitary hormone deficiency, combined 2	
NM_006261.4(PROP1):c.217C>T (p.Arg73Cys)	PROP1	5	177994231	Pituitary hormone deficiency, combined 2	
NM_006261.4(PROP1):c.295C>T (p.Arg99Ter)	PROP1	5	177994153	Pituitary hormone deficiency, combined 2	
NM_006261.4(PROP1):c.296G>A (p.Arg99Gln)	PROP1	5	177994152	Pituitary hormone deficiency, combined 2	
NM_006261.4(PROP1):c.582G>A (p.Trp194Ter)	PROP1	5	177992808	Pituitary hormone deficiency, combined 2	
NM_178138.5(LHX3):c.629C>T (p.Ala210Val)	LHX3	9	136198798	Pituitary hormone deficiency, combined 3	
NM_178138.5(LHX3):c.672G>A (p.Trp224Ter)	LHX3	9	136198755	Pituitary hormone deficiency, combined 3	
NM_033343.3(LHX4):c.250C>T (p.Arg84Cys)	LHX4	1	180266393	Pituitary hormone deficiency, combined 4	
NM_000301.3(PLG):c.704G>A (p.Arg235His)	PLG	6	160716680	Plasminogen deficiency, type I	
NM_000301.3(PLG):c.1848G>A (p.Trp616Ter)	PLG	6	160738583	Plasminogen deficiency, type I	
NM_001001547.2(CD36):c.268C>T (p.Pro90Ser)	CD36	7	80656687	Platelet glycoprotein IV deficiency	
NM_016363.5(GP6):c.524G>A (p.Ser175Asn)	GP6	19	55027664	Platelet-type bleeding disorder 11	
NM_016363.5(GP6):c.479G>A (p.Trp160Ter)	GP6	19	55027709	Platelet-type bleeding disorder 11	
NM_001060.5(TBXA2R):c.910G>A (p.Asp304Asn)	TBXA2R	19	3595810	Platelet-type bleeding disorder 13, susceptibility to	
NM_001130004.1(ACTN1):c.313G>A (p.Val105Ile)	ACTN1	14	68921033	Platelet-type bleeding disorder 15	
NM_001130004.1(ACTN1):c.2255G>A (p.Arg752Gln)	ACTN1	14	68879987	Platelet-type bleeding disorder 15	
NM_001130004.1(ACTN1):c.137G>A (p.Arg46Gln)	ACTN1	14	68925641	Platelet-type bleeding disorder 15	
NM_001130004.1(ACTN1):c.2212C>T (p.Arg738Trp)	ACTN1	14	68880030	Platelet-type bleeding disorder 15	
NM_001130004.1(ACTN1):c.673G>A (p.Glu225Lys)	ACTN1	14	68904658	Platelet-type bleeding disorder 15	
NM_000419.4(ITGA2B):c.3077G>A (p.Arg1026Gln)	ITGA2B	17	44372407	Platelet-type bleeding disorder 16	
NM_000419.4(ITGA2B):c.3076C>T (p.Arg1026Trp)	ITGA2B	17	44372408	Platelet-type bleeding disorder 16	
NM_004188.6(GFI1B):c.859C>T (p.Gln287Ter)	GFI1B	9	132990916	Platelet-type bleeding disorder 17	
NM_001129820.1(SLFN14):c.667C>T (p.Arg223Trp)	SLFN14	17	35557396	Platelet-type bleeding disorder 20	
NM_022788.4(P2RY12):c.767G>A (p.Arg256Gln)	P2RY12	3	151338079	Platelet-type bleeding disorder 8	
NM_022788.4(P2RY12):c.793C>T (p.Arg265Trp)	P2RY12	3	151338053	Platelet-type bleeding disorder 8	
NM_144606.5(FLCN):c.779G>A (p.Trp260Ter)	FLCN	17	17222501	Pneumothorax, primary spontaneous	
NM_198947.3(FAM111B):c.1883G>A (p.Ser628Asn)	FAM111B	11	59125980	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis	
NM_152692.4(C1GALT1C1):c.202C>T (p.Arg68Ter)	C1GALT1C1	X	120626965	Polyagglutinable erythrocyte syndrome	
NM_152692.4(C1GALT1C1):c.454G>A (p.Glu152Lys)	C1GALT1C1	X	120626713	Polyagglutinable erythrocyte syndrome	
NM_001282227.1(ADA2):c.380G>A (p.Arg127Gln)	ADA2	22	17207107	Polyarteritis nodosa	
NM_001282227.1(ADA2):c.13G>A (p.Gly5Arg)	ADA2	22	17209539	Polyarteritis nodosa	
NM_001282227.1(ADA2):c.626C>T (p.Pro209Leu)	ADA2	22	17203564	Polyarteritis nodosa	
NM_000297.3(PKD2):c.1390C>T (p.Arg464Ter)	PKD2	4	88046712	Polycystic kidney disease 2	
NM_000297.3(PKD2):c.1139G>A (p.Trp380Ter)	PKD2	4	88043277	Polycystic kidney disease 2	
NM_000297.3(PKD2):c.2224C>T (p.Arg742Ter)	PKD2	4	88065479	Polycystic kidney disease 2	
NM_000297.3(PKD2):c.1213C>T (p.Gln405Ter)	PKD2	4	88043351	Polycystic kidney disease 2	
NM_000297.3(PKD2):c.916C>T (p.Arg306Ter)	PKD2	4	88038323	Polycystic kidney disease 2	
NM_198335.3(GANAB):c.2515C>T (p.Arg839Trp)	GANAB	11	62626633	Polycystic kidney disease 3	
NM_001009944.2(PKD1):c.12124C>T (p.Gln4042Ter)	PKD1	16	2090688	Polycystic kidney disease, adult type	
NM_001009944.2(PKD1):c.12682C>T (p.Arg4228Ter)	PKD1	16	2089957	Polycystic kidney disease, adult type	
NM_001009944.2(PKD1):c.11512C>T (p.Gln3838Ter)	PKD1	16	2091806	Polycystic kidney disease, adult type	

NM_001009944.2(PKD1):c.5764C>T (p.Gln1922Ter)	PKD1	16	2109403	Polycystic kidney disease, adult type
NM_001009944.2(PKD1):c.12420G>A (p.Trp4140Ter)	PKD1	16	2090309	Polycystic kidney disease, adult type
NM_001009944.2(PKD1):c.9829C>T (p.Arg3277Cys)	PKD1	16	2099955	Polycystic kidney disease, adult type
NM_001009944.2(PKD1):c.10583G>A (p.Trp3528Ter)	PKD1	16	2094127	Polycystic kidney disease, adult type
NM_000297.3(PKD2):c.973C>T (p.Arg325Ter)	PKD2	4	88038380	Polycystic kidney disease, autosomal dominant
NM_000297.3(PKD2):c.145C>T (p.Gln49Ter)	PKD2	4	88007878	Polycystic kidney dysplasia
NM_001009944.2(PKD1):c.5086C>T (p.Gln1696Ter)	PKD1	16	2110081	Polycystic kidney dysplasia
NM_018965.3(TREM2):c.233G>A (p.Trp78Ter)	TREM2	6	41161421	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
NM_018965.3(TREM2):c.132G>A (p.Trp44Ter)	TREM2	6	41161522	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
NM_018965.3(TREM2):c.97C>T (p.Gln33Ter)	TREM2	6	41161557	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
NM_018965.3(TREM2):c.197C>T (p.Thr66Met)	TREM2	6	41161457	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
NM_007214.4(SEC63):c.173G>A (p.Trp58Ter)	SEC63	6	107929466	Polycystic liver disease 2
NM_007214.4(SEC63):c.733+1G>A	SEC63	6	107908926	Polycystic liver disease 2
NM_022458.3(LMBR1):c.423+4909C>T	LMBR1	7	156791480	Polydactyly, preaxial II
NM_022458.3(LMBR1):c.423+4810G>A	LMBR1	7	156791579	Polydactyly, preaxial II
NM_022458.3(LMBR1):c.423+4915C>T	LMBR1	7	156791474	Polydactyly, preaxial II;Tibia, hypoplasia or aplasia of, with polydactyly;Triphalangeal thumb
NM_000383.3(AIRE):c.769C>T (p.Arg257Ter)	AIRE	21	44289773	Polyglandular autoimmune syndrome, type 1;not provided
NM_000383.3(AIRE):c.415C>T (p.Arg139Ter)	AIRE	21	44287085	Polyglandular autoimmune syndrome, type 1;not provided
NM_000383.3(AIRE):c.1616C>T (p.Pro539Leu)	AIRE	21	44297705	Polyglandular autoimmune syndrome, type 1;not provided
NM_000158.3(GBE1):c.1544G>A (p.Arg515His)	GBE1	3	81577999	Polyglucosan body disease, adult
NM_031229.3(RBCK1):c.553C>T (p.Gln185Ter)	RBCK1	20	419439	Polyglucosan body myopathy 1 with or without immunodeficiency
NM_031229.3(RBCK1):c.790C>T (p.Gln264Ter)	RBCK1	20	420904	Polyglucosan body myopathy 1 with or without immunodeficiency
NM_004130.3(GYG1):c.970C>T (p.Arg324Ter)	GYG1	3	149026850	Polyglucosan body myopathy 2
NM_004130.3(GYG1):c.749G>A (p.Trp250Ter)	GYG1	3	149024193	Polyglucosan body myopathy 2
NM_178012.4(TUBB2B):c.1249G>A (p.Asp417Asn)	TUBB2B	6	3224840	Polymicrogyria, asymmetric
NM_178012.4(TUBB2B):c.1261G>A (p.Glu421Lys)	TUBB2B	6	3224828	Polymicrogyria, asymmetric
NM_178012.4(TUBB2B):c.292G>A (p.Gly98Arg)	TUBB2B	6	3225797	Polymicrogyria, asymmetric;not provided
NM_005682.6(ADGRG1):c.112C>T (p.Arg38Trp)	ADGRG1	16	57651247	Polymicrogyria, bilateral frontoparietal
NM_005682.6(ADGRG1):c.265C>T (p.His89Tyr)	ADGRG1	16	57651400	Polymicrogyria, bilateral frontoparietal
NM_005682.6(ADGRG1):c.286C>T (p.Arg96Ter)	ADGRG1	16	57651421	Polymicrogyria, bilateral frontoparietal
NM_005682.6(ADGRG1):c.620+1G>A	ADGRG1	16	57653336	Polymicrogyria, bilateral frontoparietal
NM_005682.6(ADGRG1):c.1426C>T (p.Arg476Ter)	ADGRG1	16	57659534	Polymicrogyria, bilateral frontoparietal
NM_005682.6(ADGRG1):c.1970G>A (p.Trp657Ter)	ADGRG1	16	57663470	Polymicrogyria, bilateral frontoparietal
NM_005682.6(ADGRG1):c.10C>T (p.Gln4Ter)	ADGRG1	16	57650297	Polymicrogyria, bilateral frontoparietal
NM_005682.6(ADGRG1):c.1693C>T (p.Arg565Trp)	ADGRG1	16	57661707	Polymicrogyria, bilateral frontoparietal;not provided
NM_058004.3(PI4KA):c.2386C>T (p.Arg796Ter)	PI4KA	22	20765636	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis
NM_058004.3(PI4KA):c.5560G>A (p.Asp1854Asn)	PI4KA	22	20713292	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis
NM_015600.4(ABHD12):c.1054C>T (p.Arg352Ter)	ABHD12	20	25302322	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract
NM_015600.4(ABHD12):c.477G>A (p.Trp159Ter)	ABHD12	20	25320264	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract
NM_003384.2(VRK1):c.397C>T (p.Arg133Cys)	VRK1	14	96852853	Pontocerebellar hypoplasia type 1
NM_003384.2(VRK1):c.706G>A (p.Val236Met)	VRK1	14	96855353	Pontocerebellar hypoplasia type 1
NM_001145394.1(TSEN2):c.757G>A (p.Gly253Arg)	TSEN2	3	12516635	Pontocerebellar hypoplasia type 2B
NM_001145392.1(TSEN2):c.691C>T (p.Gln231Ter)	TSEN2	3	12503644	Pontocerebellar hypoplasia type 2B
NM_024075.4(TSEN34):c.172C>T (p.Arg58Trp)	TSEN34	19	54191536	Pontocerebellar hypoplasia type 2C
NM_016955.3(SEPSECS):c.715G>A (p.Ala239Thr)	SEPSECS	4	25152049	Pontocerebellar hypoplasia type 2D

NM_033026.5(PCLO):c.10624C>T (p.Arg3542Ter)	PCLO	7	82949964	Pontocerebellar hypoplasia type 3
NM_207346.2(TSEN54):c.736C>T (p.Gln246Ter)	TSEN54	17	75521817	Pontocerebellar hypoplasia type 4
NM_207346.2(TSEN54):c.1027C>T (p.Gln343Ter)	TSEN54	17	75522108	Pontocerebellar hypoplasia type 4
NM_002768.4(CHMP1A):c.28-13G>A	CHMP1A	16	89651659	Pontocerebellar hypoplasia type 8
NM_002768.4(CHMP1A):c.88C>T (p.Gln30Ter)	CHMP1A	16	89651586	Pontocerebellar hypoplasia type 8;not provided
NM_006831.2(CLP1):c.419G>A (p.Arg140His)	CLP1	11	57659895	Pontocerebellar hypoplasia, type 10
NM_181503.2(EXOSC8):c.5C>T (p.Ala2Val)	EXOSC8	13	37000810	Pontocerebellar hypoplasia, type 1c
NM_001128159.2(VPS53):c.1556+5G>A	VPS53	17	562498	Pontocerebellar hypoplasia, type 2e
NM_052965.3(TSEN15):c.346C>T (p.His116Tyr)	TSEN15	1	184054856	Pontocerebellar hypoplasia, type 2f
NM_001257360.1(AMPD2):c.2021G>A (p.Arg674His)	AMPD2	1	109629487	Pontocerebellar hypoplasia, type 9
NM_006147.3(IRF6):c.1177C>T (p.Gln393Ter)	IRF6	1	209789669	Popliteal pterygium syndrome
NM_006147.3(IRF6):c.251G>A (p.Arg84His)	IRF6	1	209796476	Popliteal pterygium syndrome
NM_006147.3(IRF6):c.1271C>T (p.Ser424Leu)	IRF6	1	209788553	Popliteal pterygium syndrome
NM_006147.3(IRF6):c.250C>T (p.Arg84Cys)	IRF6	1	209796477	Popliteal pterygium syndrome;not provided
NM_001845.5(COL4A1):c.3706G>A (p.Gly1236Arg)	COL4A1	13	110170583	Porencephaly 1
NM_001845.5(COL4A1):c.2245G>A (p.Gly749Ser)	COL4A1	13	110179370	Porencephaly 1
NM_001845.5(COL4A1):c.3389G>A (p.Gly1130Asp)	COL4A1	13	110174463	Porencephaly 1
NM_001846.3(COL4A2):c.3455G>A (p.Gly1152Asp)	COL4A2	13	110492070	Porencephaly 2
NM_001846.3(COL4A2):c.3110G>A (p.Gly1037Glu)	COL4A2	13	110485739	Porencephaly 2
NM_022082.3(SLC17A9):c.932G>A (p.Arg311Gln)	SLC17A9	20	62965153	Porokeratosis 8, disseminated superficial actinic type
NM_022082.3(SLC17A9):c.25C>T (p.Arg9Cys)	SLC17A9	20	62952855	Porokeratosis 8, disseminated superficial actinic type
NM_002004.3(FDPS):c.536G>A (p.Arg179Gln)	FDPS	1	155317996	Porokeratosis 9, multiple types
NM_001135822.1(FDPS):c.486+1G>A	FDPS	1	155318292	Porokeratosis 9, multiple types
NM_006556.3(PMVK):c.412C>T (p.Arg138Ter)	PMVK	1	154926384	Porokeratosis of Mibelli
NM_000431.3(MVK):c.1039+1G>A	MVK	12	109595182	Porokeratosis, disseminated superficial actinic 1
NM_000031.5(ALAD):c.397G>A (p.Gly133Arg)	ALAD	9	113390798	Porphobilinogen synthase deficiency
NM_000031.5(ALAD):c.823G>A (p.Val275Met)	ALAD	9	113389085	Porphobilinogen synthase deficiency
NM_000031.5(ALAD):c.718C>T (p.Arg240Trp)	ALAD	9	113389521	Porphobilinogen synthase deficiency
NM_000031.5(ALAD):c.820G>A (p.Ala274Thr)	ALAD	9	113389088	Porphobilinogen synthase deficiency
NM_000031.5(ALAD):c.165-11C>T	ALAD	9	113391634	Porphobilinogen synthase deficiency
NM_000374.4(UROD):c.346C>T (p.Gln116Ter)	UROD	1	45013663	Porphyria cutanea tarda
NM_003441.3(ZNF141):c.1421C>T (p.Thr474Ile)	ZNF141	4	373858	Postaxial polydactyly type A6
NM_000168.5(GLI3):c.1927C>T (p.Arg643Ter)	GLI3	7	41972513	Postaxial polydactyly, type a1/b
NM_014053.3(FLVCR1):c.721G>A (p.Ala241Thr)	FLVCR1	1	212859173	Posterior column ataxia with retinitis pigmentosa
NM_000089.3(COL1A2):c.1981G>A (p.Gly661Ser)	COL1A2	7	94418508	Postmenopausal osteoporosis
NM_000334.4(SCN4A):c.3917G>A (p.Gly1306Glu)	SCN4A	17	63943846	Potassium aggravated myotonia
NM_000334.4(SCN4A):c.1333G>A (p.Val445Met)	SCN4A	17	63964587	Potassium aggravated myotonia;not provided
NM_000506.4(F2):c.*97G>A	F2	11	46739505	Pregnancy loss, recurrent, susceptibility to, 2;Prothrombin deficiency, congenital;Thrombophilia
NM_207421.4(PADI6):c.1141C>T (p.Gln381Ter)	PADI6	1	17394041	PREIMPLANTATION EMBRYONIC LETHALITY 2
NM_207421.4(PADI6):c.970C>T (p.Gln324Ter)	PADI6	1	17392121	PREIMPLANTATION EMBRYONIC LETHALITY 2
NM_207421.4(PADI6):c.1618G>A (p.Gly540Arg)	PADI6	1	17395663	PREIMPLANTATION EMBRYONIC LETHALITY 2
NM_000892.4(KLKB1):c.337C>T (p.Arg113Ter)	KLKB1	4	186236789	Prekallikrein deficiency
NM_000892.4(KLKB1):c.1205G>A (p.Trp402Ter)	KLKB1	4	186252077	Prekallikrein deficiency
NM_000892.4(KLKB1):c.1643G>A (p.Cys548Tyr)	KLKB1	4	186257283	Prekallikrein deficiency
NM_000892.4(KLKB1):c.367G>A (p.Gly123Arg)	KLKB1	4	186236819	Prekallikrein deficiency

NM_000892.4(KLKB1):c.428G>A (p.Ser143Asn)	KLKB1	4	186236880	Prekallikrein deficiency	
NM_032485.5(MCM8):c.1954-1G>A	MCM8	20	5985921	Premature ovarian failure 10	
NM_001143763.1(SYCE1):c.721C>T (p.Gln241Ter)	SYCE1	10	133555706	Premature ovarian failure 12	
NM_024921.3(POF1B):c.986G>A (p.Arg329Gln)	POF1B	X	85308188	Premature ovarian failure 2b	
NM_005448.2(BMP15):c.226C>T (p.Arg76Cys)	BMP15	X	50911009	Premature ovarian failure 4	
NM_005448.2(BMP15):c.631C>T (p.Gln211Ter)	BMP15	X	50916059	Premature ovarian failure 4	
NM_001080413.3(NOBOX):c.1064G>A (p.Arg355His)	NOBOX	7	144399847	Premature ovarian failure 5	
NM_001080413.3(NOBOX):c.907C>T (p.Arg303Ter)	NOBOX	7	144400250	Premature ovarian failure 5	
NM_001017975.4(HFM1):c.2206G>A (p.Gly736Ser)	HFM1	1	91350738	Premature ovarian failure 9	
NM_000720.3(CACNA1D):c.1208G>A (p.Gly403Asp)	CACNA1D	3	53673804	Primary aldosteronism, seizures, and neurologic abnormalities;not provided	
NM_007174.2(CIT):c.412C>T (p.Gln138Ter)	CIT	12	119857525	Primary autosomal recessive microcephaly	
NM_024596.4(MCPH1):c.215C>T (p.Ser72Leu)	MCPH1	8	6414865	Primary autosomal recessive microcephaly 1	
NM_024596.4(MCPH1):c.2145G>A (p.Trp715Ter)	MCPH1	8	6499860	Primary autosomal recessive microcephaly 1;not provided	
NM_022095.3(ZNF335):c.3332G>A (p.Arg1111His)	ZNF335	20	45950453	Primary autosomal recessive microcephaly 10	
NM_001145306.1(CDK6):c.589G>A (p.Ala197Thr)	CDK6	7	92671484	Primary autosomal recessive microcephaly 12	
NM_001813.2(CENPE):c.2797G>A (p.Asp933Asn)	CENPE	4	103158691	Primary autosomal recessive microcephaly 13;not provided	
NM_032793.4(MFSD2A):c.476C>T (p.Thr159Met)	MFSD2A	1	39965333	Primary autosomal recessive microcephaly 15	
NM_032793.4(MFSD2A):c.497C>T (p.Ser166Leu)	MFSD2A	1	39965490	Primary autosomal recessive microcephaly 15	
NM_032793.4(MFSD2A):c.1016C>T (p.Ser339Leu)	MFSD2A	1	39967632	Primary autosomal recessive microcephaly 15	
NM_001083961.1(WDR62):c.1408C>T (p.Gln470Ter)	WDR62	19	36083099	Primary autosomal recessive microcephaly 2	
NM_001083961.1(WDR62):c.1313G>A (p.Arg438His)	WDR62	19	36081512	Primary autosomal recessive microcephaly 2	
NM_001083961.1(WDR62):c.1531G>A (p.Asp511Asn)	WDR62	19	36083222	Primary autosomal recessive microcephaly 2	
NM_001083961.1(WDR62):c.193G>A (p.Val65Met)	WDR62	19	36058795	Primary autosomal recessive microcephaly 2	
NM_001083961.1(WDR62):c.332+1G>A	WDR62	19	36060031	Primary autosomal recessive microcephaly 2	
NM_018249.5(CDK5RAP2):c.4672C>T (p.Arg1558Ter)	CDK5RAP2	9	120408401	Primary autosomal recessive microcephaly 3	
NM_018249.5(CDK5RAP2):c.5227C>T (p.Gln1743Ter)	CDK5RAP2	9	120402886	Primary autosomal recessive microcephaly 3	
NM_018249.5(CDK5RAP2):c.4441C>T (p.Arg1481Ter)	CDK5RAP2	9	120409290	Primary autosomal recessive microcephaly 3	
NM_018249.5(CDK5RAP2):c.4005-1G>A	CDK5RAP2	9	120419961	Primary autosomal recessive microcephaly 3	
NM_018249.5(CDK5RAP2):c.4207C>T (p.Arg1403Ter)	CDK5RAP2	9	120415130	Primary autosomal recessive microcephaly 3	
NM_018136.4(ASPM):c.3978G>A (p.Trp1326Ter)	ASPM	1	197117876	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.349C>T (p.Arg117Ter)	ASPM	1	197144049	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.2389C>T (p.Arg797Ter)	ASPM	1	197133380	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.1990C>T (p.Gln664Ter)	ASPM	1	197139803	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.2938C>T (p.Arg980Ter)	ASPM	1	197125190	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.2967G>A (p.Trp989Ter)	ASPM	1	197125161	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.3055C>T (p.Arg1019Ter)	ASPM	1	197125073	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.3082G>A (p.Gly1028Arg)	ASPM	1	197125046	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.3741+1G>A	ASPM	1	197122158	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.3811C>T (p.Arg1271Ter)	ASPM	1	197121974	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.4074G>A (p.Trp1358Ter)	ASPM	1	197105177	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.4795C>T (p.Arg1599Ter)	ASPM	1	197104456	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.577C>T (p.Gln193Ter)	ASPM	1	197143675	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.6232C>T (p.Arg2078Ter)	ASPM	1	197103019	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.7894C>T (p.Gln2632Ter)	ASPM	1	197101357	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.8668C>T (p.Gln2890Ter)	ASPM	1	197100583	Primary autosomal recessive microcephaly 5	

NM_018136.4(ASPM):c.9319C>T (p.Arg3107Ter)	ASPM	1	197092032	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.10168C>T (p.Arg3390Ter)	ASPM	1	197086966	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.2791C>T (p.Arg931Ter)	ASPM	1	197128635	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.3830G>A (p.Trp1277Ter)	ASPM	1	197121955	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.5233C>T (p.Arg1745Ter)	ASPM	1	197104018	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.7612C>T (p.Gln2538Ter)	ASPM	1	197101639	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.7825C>T (p.Gln2609Ter)	ASPM	1	197101426	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.8017C>T (p.Gln2673Ter)	ASPM	1	197101234	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.8704C>T (p.Gln2902Ter)	ASPM	1	197100547	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.8903G>A (p.Trp2968Ter)	ASPM	1	197096082	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.9091C>T (p.Arg3031Ter)	ASPM	1	197093255	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.10060C>T (p.Arg3354Ter)	ASPM	1	197088357	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.8829G>A (p.Trp2943Ter)	ASPM	1	197096156	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.5467C>T (p.Gln1823Ter)	ASPM	1	197103784	Primary autosomal recessive microcephaly 5	
NM_018136.4(ASPM):c.9178C>T (p.Gln3060Ter)	ASPM	1	197093168	Primary autosomal recessive microcephaly 5;not provided	
NM_018136.4(ASPM):c.9190C>T (p.Arg3064Ter)	ASPM	1	197093156	Primary autosomal recessive microcephaly 5;not provided	
NM_018136.4(ASPM):c.9697C>T (p.Arg3233Ter)	ASPM	1	197090328	Primary autosomal recessive microcephaly 5;not provided	
NM_018136.4(ASPM):c.9730C>T (p.Arg3244Ter)	ASPM	1	197090295	Primary autosomal recessive microcephaly 5;not provided	
NM_018136.4(ASPM):c.1138C>T (p.Gln380Ter)	ASPM	1	197143114	Primary autosomal recessive microcephaly 5;not provided	
NM_018136.4(ASPM):c.9454C>T (p.Arg3152Ter)	ASPM	1	197091032	Primary autosomal recessive microcephaly 5;not provided	
NM_018136.4(ASPM):c.9910C>T (p.Arg3304Ter)	ASPM	1	197090004	Primary autosomal recessive microcephaly 5;not provided	
NM_018451.4(CENPJ):c.3448C>T (p.Gln1150Ter)	CENPJ	13	24885305	Primary autosomal recessive microcephaly 6	
NM_003035.2(STIL):c.3715C>T (p.Gln1239Ter)	STIL	1	47251285	Primary autosomal recessive microcephaly 7	
NM_003035.2(STIL):c.2826+1G>A	STIL	1	47262902	Primary autosomal recessive microcephaly 7	
NM_003035.2(STIL):c.266_453del	STIL	1	47301556	Primary autosomal recessive microcephaly 7	
NM_001194998.1(CEP152):c.2959C>T (p.Arg987Ter)	CEP152	15	48756289	Primary autosomal recessive microcephaly 9	
NM_001369.2(DNAH5):c.8998C>T (p.Arg3000Ter)	DNAH5	5	13777309	Primary ciliary dyskinesia	
NM_001369.2(DNAH5):c.9502C>T (p.Arg3168Ter)	DNAH5	5	13770852	Primary ciliary dyskinesia	
NM_017950.3(CCDC40):c.961C>T (p.Arg321Ter)	CCDC40	17	80050085	Primary ciliary dyskinesia	
NM_001369.2(DNAH5):c.5281C>T (p.Arg1761Ter)	DNAH5	5	13841895	Primary ciliary dyskinesia	
NM_001369.2(DNAH5):c.6249G>A (p.Met2083Ile)	DNAH5	5	13830026	Primary ciliary dyskinesia	
NM_001277115.1(DNAH11):c.4333C>T (p.Arg1445Ter)	DNAH11	7	21619178	Primary ciliary dyskinesia	
NM_001277115.1(DNAH11):c.6244C>T (p.Arg2082Ter)	DNAH11	7	21702773	Primary ciliary dyskinesia	
NM_001369.2(DNAH5):c.10384C>T (p.Gln3462Ter)	DNAH5	5	13758881	Primary ciliary dyskinesia	
NM_001369.2(DNAH5):c.8029C>T (p.Arg2677Ter)	DNAH5	5	13793710	Primary ciliary dyskinesia	
NM_001369.2(DNAH5):c.7034G>A (p.Trp2345Ter)	DNAH5	5	13814801	Primary ciliary dyskinesia	
NM_001277115.1(DNAH11):c.3122G>A (p.Trp1041Ter)	DNAH11	7	21660797	Primary ciliary dyskinesia	
NM_001277115.1(DNAH11):c.4438C>T (p.Arg1480Ter)	DNAH11	7	21620016	Primary ciliary dyskinesia	
NM_001277115.1(DNAH11):c.4621C>T (p.Arg1541Ter)	DNAH11	7	21635991	Primary ciliary dyskinesia	
NM_001277115.1(DNAH11):c.10324C>T (p.Gln3442Ter)	DNAH11	7	21808041	Primary ciliary dyskinesia	
NM_012144.3(DNAI1):c.1307G>A (p.Trp436Ter)	DNAI1	9	34506870	Primary ciliary dyskinesia	
NM_012144.3(DNAI1):c.1644G>A (p.Trp548Ter)	DNAI1	9	34514468	Primary ciliary dyskinesia	
NM_023036.4(DNAI2):c.1420G>A (p.Gly474Arg)	DNAI2	17	74310089	Primary ciliary dyskinesia	
NM_001369.2(DNAH5):c.13486C>T (p.Arg4496Ter)	DNAH5	5	13701289	Primary ciliary dyskinesia	
NM_001369.2(DNAH5):c.12265C>T (p.Gln4089Ter)	DNAH5	5	13721014	Primary ciliary dyskinesia	

NM_001369.2(DNAH5):c.5983C>T (p.Arg1995Ter)	DNAH5	5	13830675	Primary ciliary dyskinesia
NM_001369.2(DNAH5):c.11725C>T (p.Arg3909Ter)	DNAH5	5	13735167	Primary ciliary dyskinesia
NM_001369.2(DNAH5):c.13331G>A (p.Trp4444Ter)	DNAH5	5	13708130	Primary ciliary dyskinesia
NM_001369.2(DNAH5):c.8465G>A (p.Trp2822Ter)	DNAH5	5	13788898	Primary ciliary dyskinesia
NM_001369.2(DNAH5):c.2533C>T (p.Gln845Ter)	DNAH5	5	13891020	Primary ciliary dyskinesia
NM_001369.2(DNAH5):c.8314C>T (p.Arg2772Ter)	DNAH5	5	13792128	Primary ciliary dyskinesia
NM_001206927.1(DNAH8):c.7866G>A (p.Trp2622Ter)	DNAH8	6	38882917	Primary ciliary dyskinesia
NM_001277115.1(DNAH11):c.10789C>T (p.Gln3597Ter)	DNAH11	7	21842641	Primary ciliary dyskinesia
NM_021147.4(CCNO):c.775C>T (p.Gln259Ter)	CCNO	5	55231653	Primary ciliary dyskinesia
NM_001010892.2(RSPH4A):c.1068G>A (p.Trp356Ter)	RSPH4A	6	116627775	Primary ciliary dyskinesia
NM_001277115.1(DNAH11):c.13075C>T (p.Arg4359Ter)	DNAH11	7	21899361	Primary ciliary dyskinesia
NM_178452.5(DNAAF1):c.190C>T (p.Gln64Ter)	DNAAF1	16	84149072	Primary ciliary dyskinesia
NM_213607.2(CCDC103):c.115C>T (p.Gln39Ter)	CCDC103	17	44901113	Primary ciliary dyskinesia
NM_023036.4(DNAI2):c.876G>A (p.Trp292Ter)	DNAI2	17	74301057	Primary ciliary dyskinesia
NM_023036.4(DNAI2):c.883C>T (p.Arg295Ter)	DNAI2	17	74301064	Primary ciliary dyskinesia
NM_023036.4(DNAI2):c.1516C>T (p.Arg506Ter)	DNAI2	17	74312024	Primary ciliary dyskinesia
NM_000328.2(RPGR):c.1366C>T (p.Gln456Ter)	RPGR	X	38297332	Primary ciliary dyskinesia
NM_018076.4(ARMC4):c.2495+1G>A	ARMC4	10	27935009	Primary ciliary dyskinesia 23
NM_018076.4(ARMC4):c.2219G>A (p.Trp740Ter)	ARMC4	10	27936759	Primary ciliary dyskinesia 23
NM_018076.4(ARMC4):c.1969C>T (p.Gln657Ter)	ARMC4	10	27940580	Primary ciliary dyskinesia 23
NM_080860.3(RSPH1):c.308G>A (p.Gly103Asp)	RSPH1	21	42486428	Primary ciliary dyskinesia 24
NM_080860.3(RSPH1):c.281G>A (p.Trp94Ter)	RSPH1	21	42486455	Primary ciliary dyskinesia 24
NM_130810.3(DNAAF4):c.808C>T (p.Arg270Ter)	DNAAF4	15	55439557	Primary ciliary dyskinesia;Primary ciliary dyskinesia 25
NM_000364.3(TNNT2):c.422G>A (p.Arg141Gln)	TNNT2	1	201364365	Primary dilated cardiomyopathy
NM_000363.4(TNNI3):c.544G>A (p.Glu182Lys)	TNNI3	19	55154035	Primary dilated cardiomyopathy;not provided
NM_001267550.2(TTN):c.71602C>T (p.Arg23868Ter)	TTN	2	178574530	Primary dilated cardiomyopathy;not provided
NM_170707.3(LMNA):c.961C>T (p.Arg321Ter)	LMNA	1	156135925	Primary dilated cardiomyopathy;not provided
NM_001256850.1(TTN):c.66398G>A (p.Trp22133Ter)	TTN	2	178574811	Primary dilated cardiomyopathy;not provided
NM_001256850.1(TTN):c.63526C>T (p.Arg21176Ter)	TTN	2	178578066	Primary dilated cardiomyopathy;not provided
NM_001256850.1(TTN):c.75793C>T (p.Arg25265Ter)	TTN	2	178565416	Primary dilated cardiomyopathy;not provided
NM_001001430.2(TNNT2):c.613C>T (p.Arg205Trp)	TNNT2	1	201361989	Primary dilated cardiomyopathy;not provided
NM_001256850.1(TTN):c.94066+1G>A	TTN	2	178538945	Primary dilated cardiomyopathy;not provided
NM_001256850.1(TTN):c.58102C>T (p.Arg19368Ter)	TTN	2	178588700	Primary dilated cardiomyopathy;not provided
NM_001256850.1(TTN):c.44423-1G>A	TTN	2	178613938	Primary dilated cardiomyopathy;not provided
NM_002977.3(SCN9A):c.2572C>T (p.Leu858Phe)	SCN9A	2	166277252	Primary erythromelalgia
NM_000256.3(MYBPC3):c.1273C>T (p.Gln425Ter)	MYBPC3	11	47343099	Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.3233G>A (p.Trp1078Ter)	MYBPC3	11	47333291	Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.3293G>A (p.Trp1098Ter)	MYBPC3	11	47333231	Primary familial hypertrophic cardiomyopathy
NM_000364.3(TNNT2):c.842+1G>A	TNNT2	1	201359622	Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.3331-1G>A	MYBPC3	11	47332974	Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.979C>T (p.Gln327Ter)	MYBPC3	11	47346318	Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.3335G>A (p.Trp1112Ter)	MYBPC3	11	47332969	Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.2747G>A (p.Trp916Ter)	MYBPC3	11	47335200	Primary familial hypertrophic cardiomyopathy
NM_000256.3(MYBPC3):c.1924C>T (p.Gln642Ter)	MYBPC3	11	47341006	Primary familial hypertrophic cardiomyopathy
NM_000257.3(MYH7):c.2198G>A (p.Gly733Glu)	MYH7	14	23425783	Primary familial hypertrophic cardiomyopathy

NM_000256.3(MYBPC3):c.484C>T (p.Gln162Ter)	MYBPC3	11	47350035	Primary familial hypertrophic cardiomyopathy
NM_000258.2(MYL3):c.281G>A (p.Arg94His)	MYL3	3	46860702	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.1458-1G>A	MYBPC3	11	47342745	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.1897+1G>A	MYBPC3	11	47341137	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.2048G>A (p.Trp683Ter)	MYBPC3	11	47339670	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3181C>T (p.Gln1061Ter)	MYBPC3	11	47333566	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3627+1G>A	MYBPC3	11	47332565	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3815-1G>A	MYBPC3	11	47331882	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.613C>T (p.Gln205Ter)	MYBPC3	11	47349815	Primary familial hypertrophic cardiomyopathy;not provided
NM_001001430.2(TNNT2):c.244G>A (p.Gly82Arg)	TNNT2	1	201365630	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.2920C>T (p.Gln974Ter)	MYBPC3	11	47333996	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3190+1G>A	MYBPC3	11	47333556	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.1090+1G>A	MYBPC3	11	47346206	Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.3346G>A (p.Glu1116Lys)	MYH7	14	23420225	Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.1012G>A (p.Val338Met)	MYH7	14	23429901	Primary familial hypertrophic cardiomyopathy;not provided
NM_000257.3(MYH7):c.596C>T (p.Ala199Val)	MYH7	14	23431804	Primary familial hypertrophic cardiomyopathy;not provided
NM_000256.3(MYBPC3):c.3641G>A (p.Trp1214Ter)	MYBPC3	11	47332245	Primary familial hypertrophic cardiomyopathy;not provided
NM_000030.2(AGXT):c.245G>A (p.Gly82Glu)	AGXT	2	240869249	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.121G>A (p.Gly41Arg)	AGXT	2	240868986	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.697C>T (p.Arg233Cys)	AGXT	2	240875125	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.698G>A (p.Arg233His)	AGXT	2	240875126	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.738G>A (p.Trp246Ter)	AGXT	2	240875166	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.466G>A (p.Gly156Arg)	AGXT	2	240871391	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.560C>T (p.Ser187Phe)	AGXT	2	240873014	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.106C>T (p.Arg36Cys)	AGXT	2	240868971	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.346G>A (p.Gly116Arg)	AGXT	2	240869350	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.568G>A (p.Gly190Arg)	AGXT	2	240873022	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.653C>T (p.Ser218Leu)	AGXT	2	240874035	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.737G>A (p.Trp246Ter)	AGXT	2	240875165	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.1049G>A (p.Gly350Asp)	AGXT	2	240878128	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.28C>T (p.Pro10Ser)	AGXT	2	240868893	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.107G>A (p.Arg36His)	AGXT	2	240868972	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.122G>A (p.Gly41Glu)	AGXT	2	240868987	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.125G>A (p.Gly42Glu)	AGXT	2	240868990	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.130C>T (p.Gln44Ter)	AGXT	2	240868995	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.139G>A (p.Gly47Arg)	AGXT	2	240869004	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.166-1G>A	AGXT	2	240869169	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.175G>A (p.Glu59Lys)	AGXT	2	240869179	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.205C>T (p.Gln69Ter)	AGXT	2	240869209	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.242C>T (p.Ser81Leu)	AGXT	2	240869246	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.283G>A (p.Glu95Lys)	AGXT	2	240869287	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.323G>A (p.Trp108Ter)	AGXT	2	240869327	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.331C>T (p.Arg111Ter)	AGXT	2	240869335	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.332G>A (p.Arg111Gln)	AGXT	2	240869336	Primary hyperoxaluria, type I
NM_000030.2(AGXT):c.353G>A (p.Arg118His)	AGXT	2	240869357	Primary hyperoxaluria, type I

NM_000030.2(AGXT):c.364C>T (p.Arg122Ter)	AGXT	2	240870649	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.409C>T (p.Gln137Ter)	AGXT	2	240870694	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.473C>T (p.Ser158Leu)	AGXT	2	240871398	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.481G>A (p.Gly161Ser)	AGXT	2	240871406	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.518G>A (p.Cys173Tyr)	AGXT	2	240871443	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.525-1G>A	AGXT	2	240872978	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.533G>A (p.Cys178Tyr)	AGXT	2	240872987	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.547G>A (p.Asp183Asn)	AGXT	2	240873001	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.595G>A (p.Gly199Ser)	AGXT	2	240873049	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.680+1G>A	AGXT	2	240874063	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.614C>T (p.Ser205Leu)	AGXT	2	240873996	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.646G>A (p.Gly216Arg)	AGXT	2	240874028	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.753G>A (p.Trp251Ter)	AGXT	2	240875181	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.776+1G>A (p.Met259fs)	AGXT	2	240875205	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.844C>T (p.Gln282Ter)	AGXT	2	240876002	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.846+1G>A	AGXT	2	240876005	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.907C>T (p.Gln303Ter)	AGXT	2	240877597	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.922C>T (p.Gln308Ter)	AGXT	2	240877612	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.943-1G>A	AGXT	2	240878021	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.956C>T (p.Pro319Leu)	AGXT	2	240878035	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.996G>A (p.Trp332Ter)	AGXT	2	240878075	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.1045G>A (p.Gly349Ser)	AGXT	2	240878124	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.1071+1G>A	AGXT	2	240878151	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.1079G>A (p.Arg360Gln)	AGXT	2	240878721	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.1102G>A (p.Ala368Thr)	AGXT	2	240878744	Primary hyperoxaluria, type I	
NM_000030.2(AGXT):c.508G>A (p.Gly170Arg)	AGXT	2	240871433	Primary hyperoxaluria, type I;not provided	
NM_012203.1(GRHPR):c.295C>T (p.Arg99Ter)	GRHPR	9	37426545	Primary hyperoxaluria, type II	
NM_012203.1(GRHPR):c.904C>T (p.Arg302Cys)	GRHPR	9	37436699	Primary hyperoxaluria, type II	
NM_012203.1(GRHPR):c.337G>A (p.Glu113Lys)	GRHPR	9	37426587	Primary hyperoxaluria, type II	
NM_012203.1(GRHPR):c.102G>A (p.Trp34Ter)	GRHPR	9	37424863	Primary hyperoxaluria, type II	
NM_012203.1(GRHPR):c.478G>A (p.Gly160Arg)	GRHPR	9	37428557	Primary hyperoxaluria, type II	
NM_012203.1(GRHPR):c.494G>A (p.Gly165Asp)	GRHPR	9	37429732	Primary hyperoxaluria, type II	
NM_012203.1(GRHPR):c.735-1G>A	GRHPR	9	37432007	Primary hyperoxaluria, type II	
NM_012203.1(GRHPR):c.905G>A (p.Arg302His)	GRHPR	9	37436700	Primary hyperoxaluria, type II	
NM_138413.3(HOGA1):c.289C>T (p.Arg97Cys)	HOGA1	10	97598852	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.134C>T (p.Pro45Leu)	HOGA1	10	97584837	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.208C>T (p.Arg70Ter)	HOGA1	10	97584911	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.227G>A (p.Gly76Asp)	HOGA1	10	97598790	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.337G>A (p.Glu113Lys)	HOGA1	10	97598900	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.346C>T (p.Gln116Ter)	HOGA1	10	97599094	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.569C>T (p.Pro190Leu)	HOGA1	10	97599780	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.733G>A (p.Val245Ile)	HOGA1	10	97601889	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.763C>T (p.Arg255Ter)	HOGA1	10	97601919	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.834G>A (p.Ala278=)	HOGA1	10	97601990	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.839C>T (p.Thr280Ile)	HOGA1	10	97611514	Primary hyperoxaluria, type III	

NM_138413.3(HOGA1):c.907C>T (p.Arg303Cys)	HOGA1	10	97611582	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.973G>A (p.Gly325Ser)	HOGA1	10	97611648	Primary hyperoxaluria, type III	
NM_138413.3(HOGA1):c.107C>T (p.Ala36Val)	HOGA1	10	97584810	Primary hyperoxaluria;Primary hyperoxaluria, type III	
NM_000315.3(PTH):c.247C>T (p.Arg83Ter)	PTH	11	13492506	Primary hyperparathyroidism	
NM_004064.4(CDKN1B):c.-80C>T	CDKN1B	12	12717760	Primary hyperparathyroidism	
NM_005630.2(SLCO2A1):c.764G>A (p.Gly255Glu)	SLCO2A1	3	133951305	Primary hypertrophic osteoarthropathy, autosomal recessive 2	
NM_006580.3(CLDN16):c.445C>T (p.Arg149Ter)	CLDN16	3	190404779	Primary hypomagnesemia	
NM_006580.3(CLDN16):c.715G>A (p.Gly239Arg)	CLDN16	3	190408436	Primary hypomagnesemia	
NM_006580.3(CLDN16):c.571G>A (p.Gly191Arg)	CLDN16	3	190404905	Primary hypomagnesemia	
NM_006580.3(CLDN16):c.593G>A (p.Gly198Asp)	CLDN16	3	190408314	Primary hypomagnesemia	
NM_006580.3(CLDN16):c.698G>A (p.Gly233Asp)	CLDN16	3	190408419	Primary hypomagnesemia	
NM_006580.3(CLDN16):c.704C>T (p.Ser235Phe)	CLDN16	3	190408425	Primary hypomagnesemia	
NM_006580.3(CLDN16):c.350G>A (p.Trp117Ter)	CLDN16	3	190402362	Primary hypomagnesemia	
NM_003999.2(OSMR):c.2081C>T (p.Pro694Leu)	OSMR	5	38925240	Primary localized cutaneous amyloidosis 1;not provided	
NM_014550.3(CARD10):c.1210C>T (p.Arg404Trp)	CARD10	22	37506365	Primary open angle glaucoma	
NM_014550.3(CARD10):c.2485C>T (p.Arg829Trp)	CARD10	22	37492794	Primary open angle glaucoma	
NM_014550.3(CARD10):c.1024G>A (p.Val342Met)	CARD10	22	37508568	Primary open angle glaucoma	
NM_014550.3(CARD10):c.983C>T (p.Ala328Val)	CARD10	22	37508609	Primary open angle glaucoma	
NM_014550.3(CARD10):c.635G>A (p.Arg212His)	CARD10	22	37516037	Primary open angle glaucoma	
NM_000261.1(MYOC):c.734G>A (p.Cys245Tyr)	MYOC	1	171636706	Primary open angle glaucoma juvenile onset 1	
NM_000261.1(MYOC):c.754G>A (p.Gly252Arg)	MYOC	1	171636686	Primary open angle glaucoma juvenile onset 1	
NM_000261.1(MYOC):c.1109C>T (p.Pro370Leu)	MYOC	1	171636331	Primary open angle glaucoma juvenile onset 1;not provided	
NM_000261.1(MYOC):c.1099G>A (p.Gly367Arg)	MYOC	1	171636341	Primary open angle glaucoma juvenile onset 1;not provided	
NM_000261.1(MYOC):c.1102C>T (p.Gln368Ter)	MYOC	1	171636338	Primary open angle glaucoma;Primary open angle glaucoma juvenile onset 1	
NM_001204.6(BMPR2):c.2695C>T (p.Arg899Ter)	BMPR2	2	202556360	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.1040G>A (p.Cys347Tyr)	BMPR2	2	202530866	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.1471C>T (p.Arg491Trp)	BMPR2	2	202552773	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.2617C>T (p.Arg873Ter)	BMPR2	2	202556282	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.1472G>A (p.Arg491Gln)	BMPR2	2	202552774	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.994C>T (p.Arg332Ter)	BMPR2	2	202530820	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.439C>T (p.Arg147Ter)	BMPR2	2	202513739	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.16C>T (p.Gln6Ter)	BMPR2	2	202377490	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.1469C>T (p.Ala490Val)	BMPR2	2	202552771	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.1398G>A (p.Trp466Ter)	BMPR2	2	202542432	Primary pulmonary hypertension	
NM_001204.6(BMPR2):c.961C>T (p.Arg321Ter)	BMPR2	2	202520195	Primary pulmonary hypertension	
NM_001127217.2(SMAD9):c.880C>T (p.Arg294Ter)	SMAD9	13	36865660	Primary pulmonary hypertension 2;not provided	
NM_002246.2(KCNK3):c.289G>A (p.Gly97Arg)	KCNK3	2	26727672	Primary pulmonary hypertension 4	
NM_002246.2(KCNK3):c.544G>A (p.Glu182Lys)	KCNK3	2	26727927	Primary pulmonary hypertension 4	
NM_001204.6(BMPR2):c.1128+1G>A	BMPR2	2	202530955	Primary pulmonary hypertension;not provided	
NM_002246.2(KCNK3):c.608G>A (p.Gly203Asp)	KCNK3	2	26727991	Primary pulmonary hypertension;Primary pulmonary hypertension 4	
NM_001204.6(BMPR2):c.631C>T (p.Arg211Ter)	BMPR2	2	202518831	Primary pulmonary hypertension;Pulmonary hypertension, primary, dexfenfluramine-associated;not provided	
NM_015426.4(POC1A):c.241C>T (p.Arg81Ter)	POC1A	3	52149850	Primordial dwarfism;Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	
NM_001164342.2(ZBTB20):c.1802C>T (p.Thr601Ile)	ZBTB20	3	114350276	Primrose syndrome	
NM_001164342.2(ZBTB20):c.1861C>T (p.Leu621Phe)	ZBTB20	3	114339370	Primrose syndrome	

NM_001164342.2(ZBTB20):c.1876G>A (p.Val626Met)	ZBTB20	3	114339355	Primrose syndrome
NM_001164342.2(ZBTB20):c.1786C>T (p.His596Tyr)	ZBTB20	3	114350292	Primrose syndrome
NM_002936.4(RNASEH1):c.424G>A (p.Val142Ile)	RNASEH1	2	3550458	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2
NM_002936.4(RNASEH1):c.469C>T (p.Arg157Ter)	RNASEH1	2	3550413	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2
NM_002936.4(RNASEH1):c.554C>T (p.Ala185Val)	RNASEH1	2	3549068	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2
NM_080916.2(DGUOK):c.130G>A (p.Glu44Lys)	DGUOK	2	73927040	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4
NM_021830.4(TWNK):c.1001G>A (p.Arg334Gln)	TWNK	10	100989211	Progressive external ophthalmoplegia with mitochondrial DNA deletions, digenic
NM_017636.3(TRPM4):c.19G>A (p.Glu7Lys)	TRPM4	19	49157885	Progressive familial heart block type 1B
NM_017636.3(TRPM4):c.490C>T (p.Arg164Trp)	TRPM4	19	49168301	Progressive familial heart block type 1B
NM_003742.2(ABCB11):c.1723C>T (p.Arg575Ter)	ABCB11	2	168970131	Progressive familial intrahepatic cholestasis 2
NM_003742.2(ABCB11):c.3169C>T (p.Arg1057Ter)	ABCB11	2	168932421	Progressive familial intrahepatic cholestasis 2
NM_018849.2(ABCB4):c.2211+1G>A	ABCB4	7	87423905	Progressive familial intrahepatic cholestasis 3
NM_018849.2(ABCB4):c.2833C>T (p.Gln945Ter)	ABCB4	7	87411984	Progressive familial intrahepatic cholestasis 3
NM_005603.4(ATP8B1):c.2674G>A (p.Gly892Arg)	ATP8B1	18	57661207	Progressive intrahepatic cholestasis
NM_005603.4(ATP8B1):c.1660G>A (p.Asp554Asn)	ATP8B1	18	57674993	Progressive intrahepatic cholestasis
NM_005603.4(ATP8B1):c.1367C>T (p.Thr456Met)	ATP8B1	18	57688361	Progressive intrahepatic cholestasis
NM_005603.4(ATP8B1):c.1804C>T (p.Arg602Ter)	ATP8B1	18	57674849	Progressive intrahepatic cholestasis
NM_005603.4(ATP8B1):c.279G>A (p.Ala93=)	ATP8B1	18	57706490	Progressive intrahepatic cholestasis
NM_153026.2(PRICKLE1):c.311G>A (p.Arg104Gln)	PRICKLE1	12	42469523	Progressive myoclonus epilepsy with ataxia;not provided
NM_001105.4(ACVR1):c.1057G>A (p.Gly353Asp)	ACVR1	2	157761077	Progressive myositis ossificans
NM_001105.4(ACVR1):c.982G>A (p.Gly328Arg)	ACVR1	2	157766005	Progressive myositis ossificans
NM_001105.4(ACVR1):c.617G>A (p.Arg206His)	ACVR1	2	157774114	Progressive myositis ossificans;not provided
NM_003880.3(WISP3):c.434G>A (p.Cys145Tyr)	WISP3	6	112064842	Progressive pseudorheumatoid dysplasia
NM_003880.3(WISP3):c.993G>A (p.Trp331Ter)	WISP3	6	112069548	Progressive pseudorheumatoid dysplasia
NM_198239.1(WISP3):c.643+1G>A	WISP3	6	112064998	Progressive pseudorheumatoid dysplasia
NM_002693.2(POLG):c.2870C>T (p.Ala957Val)	POLG	15	89320877	Progressive sclerosing poliodystrophy;not provided
NM_001937.4(DPT):c.544C>T (p.Arg182Cys)	DPT	1	168696611	Progressive sensorineural hearing impairment
NM_152754.2(SEMA3D):c.1738C>T (p.Pro580Ser)	SEMA3D	7	85012812	Progressive sensorineural hearing impairment
NM_016835.4(MAPT):c.2006C>T (p.Ser669Leu)	MAPT	17	46018675	Progressive supranuclear ophthalmoplegia;not provided
NM_000285.3(PEPD):c.826G>A (p.Asp276Asn)	PEPD	19	33401862	Prolidase deficiency
NM_000285.3(PEPD):c.551G>A (p.Arg184Gln)	PEPD	19	33464060	Prolidase deficiency
NM_000285.3(PEPD):c.833G>A (p.Gly278Asp)	PEPD	19	33401855	Prolidase deficiency
NM_000285.3(PEPD):c.1342G>A (p.Gly448Arg)	PEPD	19	33387892	Prolidase deficiency
NM_000285.3(PEPD):c.793C>T (p.Arg265Ter)	PEPD	19	33411697	Prolidase deficiency
NM_000285.3(PEPD):c.1234G>A (p.Glu412Lys)	PEPD	19	33388000	Prolidase deficiency
NM_000285.3(PEPD):c.605C>T (p.Ser202Phe)	PEPD	19	33464006	Prolidase deficiency
NM_017791.2(FLVCR2):c.977C>T (p.Ala326Val)	FLVCR2	14	75633653	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome
NM_016335.4(PRODH):c.1292G>A (p.Arg431His)	PRODH	22	18918451	Proline dehydrogenase deficiency;Schizophrenia 4
NM_016335.4(PRODH):c.1397C>T (p.Thr466Met)	PRODH	22	18918346	Proline dehydrogenase deficiency;Schizophrenia 4
NM_002621.2(CFP):c.298C>T (p.Arg100Trp)	CFP	X	47628207	Properdin deficiency, type II
NM_002621.2(CFP):c.481C>T (p.Arg161Ter)	CFP	X	47627564	Properdin deficiency, X-linked
NM_000532.4(PCCB):c.1228C>T (p.Arg410Trp)	PCCB	3	136327184	Propionic acidemia
NM_000532.4(PCCB):c.502G>A (p.Glu168Lys)	PCCB	3	136262024	Propionic acidemia
NM_000532.4(PCCB):c.1283C>T (p.Thr428Ile)	PCCB	3	136327239	Propionic acidemia
NM_000282.3(PCCA):c.412G>A (p.Ala138Thr)	PCCA	13	100155090	Propionic acidemia

NM_000532.4(PCCB):c.1495C>T (p.Arg499Ter)	PCCB	3	136328854	Propionic acidemia
NM_000532.4(PCCB):c.1534C>T (p.Arg512Cys)	PCCB	3	136329940	Propionic acidemia
NM_000532.4(PCCB):c.335G>A (p.Gly112Asp)	PCCB	3	136256586	Propionic acidemia
NM_000282.3(PCCA):c.1788G>A (p.Trp596Ter)	PCCA	13	100425674	Propionic acidemia
NM_000282.3(PCCA):c.232-1G>A	PCCA	13	100111992	Propionic acidemia
NM_000532.4(PCCB):c.183+5G>A	PCCB	3	136250563	Propionic acidemia
NM_000532.4(PCCB):c.493C>T (p.Arg165Trp)	PCCB	3	136262015	Propionic acidemia
NM_000532.4(PCCB):c.1210G>A (p.Glu404Lys)	PCCB	3	136327166	Propionic acidemia
NM_000532.4(PCCB):c.1540C>T (p.Arg514Ter)	PCCB	3	136329946	Propionic acidemia
NM_000282.3(PCCA):c.105+1G>A	PCCA	13	100089226	Propionic acidemia
NM_000282.3(PCCA):c.184-1G>A	PCCA	13	100111840	Propionic acidemia
NM_000282.3(PCCA):c.863G>A (p.Arg288Lys)	PCCA	13	100268732	Propionic acidemia
NM_000282.3(PCCA):c.1426C>T (p.Arg476Ter)	PCCA	13	100309905	Propionic acidemia
NM_000282.3(PCCA):c.2002G>A (p.Gly668Arg)	PCCA	13	100315529	Propionic acidemia
NM_000282.3(PCCA):c.2040G>A (p.Ala680=)	PCCA	13	100515567	Propionic acidemia
NM_000532.4(PCCB):c.184-1G>A	PCCB	3	136255855	Propionic acidemia
NM_000282.3(PCCA):c.937C>T (p.Arg313Ter)	PCCA	13	100273218	Propionic acidemia;not provided
NM_000532.4(PCCB):c.183+1G>A	PCCB	3	136250559	Propionic acidemia;not provided
NM_000532.4(PCCB):c.3G>A (p.Met1Ile)	PCCB	3	136250378	Propionic acidemia;not provided
NM_000532.4(PCCB):c.683C>T (p.Pro228Leu)	PCCB	3	136293784	Propionic acidemia;not provided
NM_000282.3(PCCA):c.425G>A (p.Gly142Asp)	PCCA	13	100157297	Propionic acidemia;not provided
NM_000439.4(PCSK1):c.1447G>A (p.Gly483Arg)	PCSK1	5	96399020	Proprotein convertase 1/3 deficiency
NM_000439.4(PCSK1):c.920C>T (p.Ser307Leu)	PCSK1	5	96410949	Proprotein convertase 1/3 deficiency
NM_006361.5(HOXB13):c.251G>A (p.Gly84Glu)	HOXB13	17	48728343	Prostate cancer susceptibility;not provided
NM_021133.3(RNASEL):c.3G>A (p.Met1Ile)	RNASEL	1	182586804	Prostate cancer, hereditary, 1
NM_001142615.2(EHBP1):c.1185+30064G>A	EHBP1	2	62904596	Prostate cancer, hereditary, 12
NM_033550.2(MAD1L1):c.175C>T (p.Arg59Cys)	MAD1L1	7	2225526	Prostate cancer, somatic
NM_004442.7(EPHB2):c.2167C>T (p.Gln723Ter)	EPHB2	1	22907980	Prostate cancer/brain cancer susceptibility
NM_004442.7(EPHB2):c.2035G>A (p.Asp679Asn)	EPHB2	1	22906853	Prostate cancer/brain cancer susceptibility
NM_020061.5(OPN1LW):c.1013G>A (p.Gly338Glu)	OPN1LW	X	154158844	Protan defect
NM_000313.3(PROS1):c.835C>T (p.Gln279Ter)	PROS1	3	93898462	Protein S deficiency
NM_000313.3(PROS1):c.1063C>T (p.Arg355Cys)	PROS1	3	93893025	Protein S deficiency
NM_014625.3(NPHS2):c.353C>T (p.Pro118Leu)	NPHS2	1	179564715	Proteinuria
NM_001127899.3(CLCN5):c.1238G>A (p.Trp413Ter)	CLCN5	X	50086551	Proteinuria, low molecular weight, with hypercalciuria and nephrocalcinosis
NM_000032.4(ALAS2):c.1642C>T (p.Gln548Ter)	ALAS2	X	55009302	Protoporphyria, erythropoietic, X-linked
NM_000095.2(COMP):c.1403G>A (p.Cys468Tyr)	COMP	19	18786051	Pseudoachondroplastic spondyloepiphyseal dysplasia syndrome
NM_000095.2(COMP):c.2156G>A (p.Gly719Asp)	COMP	19	18783125	Pseudoachondroplastic spondyloepiphyseal dysplasia syndrome
NM_000095.2(COMP):c.1586C>T (p.Thr529Ile)	COMP	19	18785755	Pseudoachondroplastic spondyloepiphyseal dysplasia syndrome
NM_000095.2(COMP):c.1747G>A (p.Glu583Lys)	COMP	19	18785063	Pseudoachondroplastic spondyloepiphyseal dysplasia syndrome
NM_000095.2(COMP):c.2155G>A (p.Gly719Ser)	COMP	19	18783126	Pseudoachondroplastic spondyloepiphyseal dysplasia syndrome
NM_024312.4(GNPTAB):c.2715+1G>A	GNPTAB	12	101764201	Pseudo-Hurler polydystrophy
NM_024312.4(GNPTAB):c.1196C>T (p.Ser399Phe)	GNPTAB	12	101770109	Pseudo-Hurler polydystrophy
NM_024312.4(GNPTAB):c.1001G>A (p.Arg334Gln)	GNPTAB	12	101770518	Pseudo-Hurler polydystrophy
NM_024312.4(GNPTAB):c.1325G>A (p.Cys442Tyr)	GNPTAB	12	101768120	Pseudo-Hurler polydystrophy
NM_024312.4(GNPTAB):c.1514G>A (p.Cys505Tyr)	GNPTAB	12	101766189	Pseudo-Hurler polydystrophy

NM_024312.4(GNPTAB):c.2866C>T (p.His956Tyr)	GNPTAB	12	101761613	Pseudo-Hurler polydystrophy
NM_024312.4(GNPTAB):c.1774G>A (p.Ala592Thr)	GNPTAB	12	101765143	Pseudo-Hurler polydystrophy
NM_005689.3(ABCB6):c.1124G>A (p.Arg375Gln)	ABCB6	2	219216027	Pseudohyperkalemia, familial, 2, due to red cell leak
NM_005689.3(ABCB6):c.1123C>T (p.Arg375Trp)	ABCB6	2	219216028	Pseudohyperkalemia, familial, 2, due to red cell leak
NM_005689.3(ABCB6):c.2168G>A (p.Arg723Gln)	ABCB6	2	219210799	Pseudohyperkalemia, familial, 2, due to red cell leak
NM_000901.4(NR3C2):c.1609C>T (p.Arg537Ter)	NR3C2	4	148435252	Pseudohypoaldosteronism type 1 autosomal dominant
NM_000901.4(NR3C2):c.1897G>A (p.Gly633Arg)	NR3C2	4	148259978	Pseudohypoaldosteronism type 1 autosomal dominant
NM_000901.4(NR3C2):c.2839C>T (p.Arg947Ter)	NR3C2	4	148081460	Pseudohypoaldosteronism type 1 autosomal dominant
NM_000901.4(NR3C2):c.2017C>T (p.Arg673Ter)	NR3C2	4	148154899	Pseudohypoaldosteronism type 1 autosomal dominant
NM_000901.4(NR3C2):c.2453C>T (p.Ser818Leu)	NR3C2	4	148152526	Pseudohypoaldosteronism type 1 autosomal dominant
NM_000336.2(SCNN1B):c.109G>A (p.Gly37Ser)	SCNN1B	16	23348708	Pseudohypoaldosteronism type 1 autosomal recessive
NM_001038.5(SCNN1A):c.1522C>T (p.Arg508Ter)	SCNN1A	12	6348981	Pseudohypoaldosteronism type 1 autosomal recessive
NM_001038.5(SCNN1A):c.1685C>T (p.Ser562Leu)	SCNN1A	12	6348198	Pseudohypoaldosteronism type 1 autosomal recessive
NM_032387.4(WNK4):c.1684G>A (p.Glu562Lys)	WNK4	17	42787485	Pseudohypoaldosteronism type 2B
NM_032387.4(WNK4):c.3553C>T (p.Arg1185Cys)	WNK4	17	42796244	Pseudohypoaldosteronism type 2B
NM_032387.4(WNK4):c.1682C>T (p.Pro561Leu)	WNK4	17	42787483	Pseudohypoaldosteronism type 2B
NM_017415.2(KLHL3):c.1193C>T (p.Ala398Val)	KLHL3	5	137638979	Pseudohypoaldosteronism type 2D
NM_017415.2(KLHL3):c.1277C>T (p.Pro426Leu)	KLHL3	5	137637338	Pseudohypoaldosteronism type 2D
NM_017415.2(KLHL3):c.1229C>T (p.Ser410Leu)	KLHL3	5	137637386	Pseudohypoaldosteronism type 2D;Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1583G>A (p.Arg528His)	KLHL3	5	137628305	Pseudohypoaldosteronism type 2D;Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.718C>T (p.Arg240Ter)	KLHL3	5	137661950	Pseudohypoaldosteronism type 2D;Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1582C>T (p.Arg528Cys)	KLHL3	5	137628306	Pseudohypoaldosteronism type 2D;Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1298G>A (p.Ser433Asn)	KLHL3	5	137637317	Pseudohypoaldosteronism type 2D;Pseudohypoaldosteronism, type 2
NM_003590.4(CUL3):c.1207-1G>A	CUL3	2	224503823	Pseudohypoaldosteronism, type 2
NM_003590.4(CUL3):c.1207-3C>T	CUL3	2	224503825	Pseudohypoaldosteronism, type 2
NM_003590.4(CUL3):c.1236G>A (p.Leu412=)	CUL3	2	224503793	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1410G>A (p.Trp470Ter)	KLHL3	5	137634077	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.753+1G>A	KLHL3	5	137661914	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1019C>T (p.Ala340Val)	KLHL3	5	137639862	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1480G>A (p.Ala494Thr)	KLHL3	5	137628408	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.430C>T (p.Gln144Ter)	KLHL3	5	137692381	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1151G>A (p.Arg384Gln)	KLHL3	5	137639021	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1292G>A (p.Arg431Gln)	KLHL3	5	137637323	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1723C>T (p.Arg575Trp)	KLHL3	5	137625765	Pseudohypoaldosteronism, type 2
NM_017415.2(KLHL3):c.1295G>A (p.Ser432Asn)	KLHL3	5	137637320	Pseudohypoaldosteronism, type 2
NM_001077488.3(GNAS):c.496C>T (p.Arg166Cys)	GNAS	20	58905443	Pseudohypoparathyroidism type 1A
NM_001077488.3(GNAS):c.695G>A (p.Arg232His)	GNAS	20	58909553	Pseudohypoparathyroidism type 1A
NM_001077488.3(GNAS):c.1177G>A (p.Glu393Lys)	GNAS	20	58910818	Pseudohypoparathyroidism type 1A;Pseudohypoparathyroidism type 1C;Pseudopseudohypoparathyroidism
NM_001077488.3(GNAS):c.347C>T (p.Pro116Leu)	GNAS	20	58903703	Pseudohypoparathyroidism type 1A;Pseudopseudohypoparathyroidism
NM_000516.5(GNAS):c.34C>T (p.Gln12Ter)	GNAS	20	58891760	Pseudohypoparathyroidism type 1A;Pseudopseudohypoparathyroidism
NM_004035.6(ACOX1):c.442C>T (p.Arg148Ter)	ACOX1	17	75957555	Pseudoneonatal adrenoleukodystrophy
NM_001039.3(SCNN1G):c.1718G>A (p.Trp573Ter)	SCNN1G	16	23215237	Pseudoprimary hyperaldosteronism
NM_000336.2(SCNN1B):c.1696C>T (p.Arg566Ter)	SCNN1B	16	23380574	Pseudoprimary hyperaldosteronism
NM_000336.2(SCNN1B):c.1847C>T (p.Pro616Leu)	SCNN1B	16	23380725	Pseudoprimary hyperaldosteronism

NM_000336.2(SCNN1B):c.1849C>T (p.Pro617Ser)	SCNN1B	16	23380727	Pseudoprimary hyperaldosteronism
NM_001077488.3(GNAS):c.775C>T (p.Arg259Trp)	GNAS	20	58909737	Pseudopseudohypoparathyroidism
NM_001171.5(ABCC6):c.3413G>A (p.Arg1138Gln)	ABCC6	16	16163086	Pseudoxanthoma elasticum
NM_001171.5(ABCC6):c.3389C>T (p.Thr1130Met)	ABCC6	16	16163110	Pseudoxanthoma elasticum
NM_001171.5(ABCC6):c.4015C>T (p.Arg1339Cys)	ABCC6	16	16154899	Pseudoxanthoma elasticum
NM_001171.5(ABCC6):c.3961G>A (p.Gly1321Ser)	ABCC6	16	16154953	Pseudoxanthoma elasticum
NM_001171.5(ABCC6):c.3412C>T (p.Arg1138Trp)	ABCC6	16	16163087	Pseudoxanthoma elasticum;not provided
NM_001171.5(ABCC6):c.3490C>T (p.Arg1164Ter)	ABCC6	16	16163009	Pseudoxanthoma elasticum;not provided
NM_001171.5(ABCC6):c.3904G>A (p.Gly1302Arg)	ABCC6	16	16135010	Pseudoxanthoma elasticum;not provided
NM_000821.6(GGCX):c.899C>T (p.Ser300Phe)	GGCX	2	85553488	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
NM_000821.6(GGCX):c.1672G>A (p.Gly558Arg)	GGCX	2	85551548	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
NM_000821.6(GGCX):c.1120C>T (p.Gln374Ter)	GGCX	2	85553267	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
NM_000821.6(GGCX):c.1426C>T (p.Arg476Cys)	GGCX	2	85552429	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
NM_000821.6(GGCX):c.1427G>A (p.Arg476His)	GGCX	2	85552428	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
NM_000821.6(GGCX):c.763G>A (p.Val255Met)	GGCX	2	85554269	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
NM_024110.4(CARD14):c.349+5G>A	CARD14	17	80182795	Psoriasis susceptibility 2
NM_024110.4(CARD14):c.349G>A (p.Gly117Ser)	CARD14	17	80182790	Psoriasis susceptibility 2;not provided
NM_024110.4(CARD14):c.424G>A (p.Glu142Lys)	CARD14	17	80183987	Psoriasis susceptibility 2;not provided
NM_000314.6(PTEN):c.445C>T (p.Gln149Ter)	PTEN	10	87933204	PTEN hamartoma tumor syndrome
NM_000314.6(PTEN):c.379G>A (p.Gly127Arg)	PTEN	10	87933138	PTEN hamartoma tumor syndrome;not provided
NM_000314.6(PTEN):c.781C>T (p.Gln261Ter)	PTEN	10	87957999	PTEN hamartoma tumor syndrome;not provided
NM_006424.2(SLC34A2):c.226C>T (p.Gln76Ter)	SLC34A2	4	25662818	PULMONARY ALVEOLAR MICROLITHIASIS
NM_000020.2(ACVRL1):c.632G>A (p.Gly211Asp)	ACVRL1	12	51914445	Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia
NM_198253.2(TERT):c.508G>A (p.Val170Met)	TERT	5	1294378	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1
NM_198253.2(TERT):c.2146G>A (p.Ala716Thr)	TERT	5	1278781	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1
NM_198253.2(TERT):c.2768C>T (p.Pro923Leu)	TERT	5	1264479	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1
NM_032957.4(RTEL1):c.1523C>T (p.Pro508Leu)	RTEL1	20	63687740	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3
NM_032957.4(RTEL1):c.2077C>T (p.Gln693Ter)	RTEL1	20	63689628	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3
NM_002582.3(PARN):c.529C>T (p.Gln177Ter)	PARN	16	14610669	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4
NM_001204.6(BMPR2):c.1297C>T (p.Gln433Ter)	BMPR2	2	202542331	Pulmonary hypertension, primary, 1, with hereditary hemorrhagic telangiectasia
NM_001204.6(BMPR2):c.545G>A (p.Gly182Asp)	BMPR2	2	202514903	Pulmonary hypertension, primary, fenfluramine-associated
NM_001089.2(ABCA3):c.128G>A (p.Arg43His)	ABCA3	16	2326201	Pulmonary Surfactant Metabolism Dysfunction
NM_000270.3(PNP):c.265G>A (p.Glu89Lys)	PNP	14	20474555	Purine-nucleoside phosphorylase deficiency
NM_000270.3(PNP):c.70C>T (p.Arg24Ter)	PNP	14	20472366	Purine-nucleoside phosphorylase deficiency
NM_000270.3(PNP):c.172C>T (p.Arg58Ter)	PNP	14	20472468	Purine-nucleoside phosphorylase deficiency
NM_173170.1(IL36RN):c.338C>T (p.Ser113Leu)	IL36RN	2	113062547	Pustular psoriasis, generalized
NM_173170.1(IL36RN):c.142C>T (p.Arg48Trp)	IL36RN	2	113062150	Pustular psoriasis, generalized
NM_012275.2(IL36RN):c.28C>T (p.Arg10Ter)	IL36RN	2	113059466	Pustular psoriasis, generalized
NM_012275.2(IL36RN):c.304C>T (p.Arg102Trp)	IL36RN	2	113062513	Pustular psoriasis, generalized
NM_000396.3(CTSK):c.721C>T (p.Arg241Ter)	CTSK	1	150799607	Pyknodysostosis
NM_000396.3(CTSK):c.830C>T (p.Ala277Val)	CTSK	1	150799228	Pyknodysostosis
NM_000396.3(CTSK):c.236G>A (p.Gly79Glu)	CTSK	1	150806109	Pyknodysostosis
NM_003014.3(SFRP4):c.694C>T	SFRP4	7	37912216	Pyle metaphyseal dysplasia
NM_003978.4(PSTPIP1):c.688G>A (p.Ala230Thr)	PSTPIP1	15	77031225	Pyogenic arthritis, pyoderma gangrenosum and acne;not provided
NM_003978.4(PSTPIP1):c.748G>A (p.Glu250Lys)	PSTPIP1	15	77032304	Pyogenic arthritis, pyoderma gangrenosum and acne;not provided

NM_018129.3(PNPO):c.685C>T (p.Arg229Trp)	PNPO	17	47946681	Pyridoxal 5'-phosphate-dependent epilepsy
NM_018129.3(PNPO):c.364_417del54	PNPO	17	47945558	Pyridoxal 5'-phosphate-dependent epilepsy
NM_018129.3(PNPO):c.520C>T (p.Gln174Ter)	PNPO	17	47945963	Pyridoxal 5'-phosphate-dependent epilepsy
NM_018129.3(PNPO):c.686G>A (p.Arg229Gln)	PNPO	17	47946682	Pyridoxal 5'-phosphate-dependent epilepsy;not provided
NM_001182.4(ALDH7A1):c.596C>T (p.Ala199Val)	ALDH7A1	5	126577133	Pyridoxine-dependent epilepsy
NM_001182.4(ALDH7A1):c.1093+1G>A	ALDH7A1	5	126555930	Pyridoxine-dependent epilepsy
NM_001182.4(ALDH7A1):c.986G>A (p.Arg329Lys)	ALDH7A1	5	126559262	Pyridoxine-dependent epilepsy
NM_001182.4(ALDH7A1):c.328C>T (p.Arg110Ter)	ALDH7A1	5	126583997	Pyridoxine-dependent epilepsy;Seizures;Ventriculomegaly;not provided
NM_000920.3(PC):c.1828G>A (p.Ala610Thr)	PC	11	66851944	Pyruvate carboxylase deficiency
NM_000920.3(PC):c.1351C>T (p.Arg451Cys)	PC	11	66863791	Pyruvate carboxylase deficiency
NM_001040716.1(PC):c.467G>A (p.Arg156Gln)	PC	11	66871335	Pyruvate carboxylase deficiency
NM_000920.3(PC):c.184C>T (p.Arg62Cys)	PC	11	66871824	Pyruvate carboxylase deficiency
NM_000920.3(PC):c.2540C>T (p.Ala847Val)	PC	11	66850398	Pyruvate carboxylase deficiency
NM_000920.3(PC):c.1892G>A (p.Arg631Gln)	PC	11	66851880	Pyruvate carboxylase deficiency;not provided
NM_000284.3(PDHA1):c.1133G>A (p.Arg378His)	PDHA1	X	19359613	Pyruvate dehydrogenase E1-alpha deficiency
NM_000284.3(PDHA1):c.943G>A (p.Asp315Asn)	PDHA1	X	19358959	Pyruvate dehydrogenase E1-alpha deficiency
NM_000284.3(PDHA1):c.863G>A (p.Arg288His)	PDHA1	X	19357683	Pyruvate dehydrogenase E1-alpha deficiency
NM_000284.3(PDHA1):c.422G>A (p.Arg141Gln)	PDHA1	X	19353085	Pyruvate dehydrogenase E1-alpha deficiency
NM_000284.3(PDHA1):c.904C>T (p.Arg302Cys)	PDHA1	X	19358920	Pyruvate dehydrogenase E1-alpha deficiency;not provided
NM_000925.3(PDHB):c.1030C>T (p.Pro344Ser)	PDHB	3	58428084	Pyruvate dehydrogenase E1-beta deficiency
NM_003477.2(PDHX):c.641+1G>A	PDHX	11	34960519	Pyruvate dehydrogenase E3-binding protein deficiency
NM_003477.2(PDHX):c.1024-1G>A	PDHX	11	34984569	Pyruvate dehydrogenase E3-binding protein deficiency
NM_003477.2(PDHX):c.742C>T (p.Gln248Ter)	PDHX	11	34966740	Pyruvate dehydrogenase E3-binding protein deficiency
NM_006859.3(LIAS):c.746G>A (p.Arg249His)	LIAS	4	39470027	Pyruvate dehydrogenase lipoic acid synthetase deficiency
NM_000298.5(PKLR):c.487C>T (p.Arg163Cys)	PKLR	1	155295457	Pyruvate kinase deficiency of red cells
NM_000298.5(PKLR):c.1151C>T (p.Thr384Met)	PKLR	1	155293556	Pyruvate kinase deficiency of red cells
NM_000298.5(PKLR):c.1436G>A (p.Arg479His)	PKLR	1	155293177	Pyruvate kinase deficiency of red cells
NM_000298.5(PKLR):c.1456C>T (p.Arg486Trp)	PKLR	1	155291918	Pyruvate kinase deficiency of red cells
NM_000298.5(PKLR):c.1529G>A (p.Arg510Gln)	PKLR	1	155291845	Pyruvate kinase deficiency of red cells;not provided
NM_005105.4(RBM8A):c.487C>T (p.Arg163Ter)	RBM8A	1	145925920	Radial aplasia-thrombocytopenia syndrome
NM_005105.4(RBM8A):c.-21G>A	RBM8A	1	145927447	Radial aplasia-thrombocytopenia syndrome;not provided
NM_004991.3(MECOM):c.2812C>T (p.Arg938Trp)	MECOM	3	169100922	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2
NM_020223.3(FAM20C):c.1645C>T (p.Arg549Trp)	FAM20C	7	259870	Raine syndrome
NM_020223.3(FAM20C):c.1136G>A (p.Gly379Glu)	FAM20C	7	255912	Raine syndrome
NM_020223.3(FAM20C):c.1446-1G>A	FAM20C	7	258645	Raine syndrome
NM_020223.3(FAM20C):c.796G>A (p.Gly266Arg)	FAM20C	7	208909	Raine syndrome
NM_020223.3(FAM20C):c.982C>T (p.Pro328Ser)	FAM20C	7	248340	Raine syndrome
NM_004260.3(RECQL4):c.806G>A (p.Trp269Ter)	RECQL4	8	144516313	Rapadilino syndrome
NM_004260.3(RECQL4):c.2476C>T (p.Arg826Ter)	RECQL4	8	144513126	Rapadilino syndrome;Rothmund-Thomson syndrome;not provided
NM_002834.4(PTPN11):c.181G>A (p.Asp61Asn)	PTPN11	12	112450361	Rasopathy;not provided
NM_030662.3(MAP2K2):c.619G>A (p.Glu207Lys)	MAP2K2	19	4101105	Rasopathy;not provided
NM_000094.3(COL7A1):c.7957G>A (p.Gly2653Arg)	COL7A1	3	48567736	Recessive dystrophic epidermolysis bullosa
NM_000094.3(COL7A1):c.8245G>A (p.Gly2749Arg)	COL7A1	3	48566719	Recessive dystrophic epidermolysis bullosa
NM_000094.3(COL7A1):c.8278G>A (p.Gly2760Arg)	COL7A1	3	48566686	Recessive dystrophic epidermolysis bullosa
NM_000094.3(COL7A1):c.6091G>A (p.Gly2031Ser)	COL7A1	3	48575428	Recessive dystrophic epidermolysis bullosa;not provided

NM_000094.3(COL7A1):c.6187C>T (p.Arg2063Trp)	COL7A1	3	48575236	Recessive dystrophic epidermolysis bullosa;not provided
NM_000094.3(COL7A1):c.7411C>T (p.Arg2471Ter)	COL7A1	3	48570304	Recessive dystrophic epidermolysis bullosa;not provided
NM_000094.3(COL7A1):c.8440C>T (p.Arg2814Ter)	COL7A1	3	48565636	Recessive dystrophic epidermolysis bullosa;not provided
NM_006214.3(PHYH):c.610G>A (p.Gly204Ser)	PHYH	10	13288428	Refsum disease, adult, 1
NM_006214.3(PHYH):c.824G>A (p.Arg275Gln)	PHYH	10	13283694	Refsum disease, adult, 1
NM_006214.3(PHYH):c.823C>T (p.Arg275Trp)	PHYH	10	13283695	Refsum disease, adult, 1;not provided
NM_000044.4(AR):c.1789G>A (p.Ala597Thr)	AR	X	67686030	Reifenstein syndrome
NM_000044.4(AR):c.2567G>A (p.Arg856His)	AR	X	67722944	Reifenstein syndrome
NM_000044.4(AR):c.2522G>A (p.Arg841His)	AR	X	67722899	Reifenstein syndrome
NM_000044.4(AR):c.4G>A (p.Glu2Lys)	AR	X	67545150	Reifenstein syndrome
NM_000044.4(AR):c.1174C>T (p.Pro392Ser)	AR	X	67546320	Reifenstein syndrome
NM_000044.4(AR):c.1823G>A (p.Arg608Gln)	AR	X	67686064	Reifenstein syndrome;not provided
NM_000358.2(TGFBI):c.1868G>A (p.Gly623Asp)	TGFBI	5	136060898	Reis-Bucklers' corneal dystrophy
NM_003638.2(ITGA8):c.1219G>A (p.Gly407Arg)	ITGA8	10	15644210	Renal adysplasia
NM_003060.3(SLC22A5):c.396G>A (p.Trp132Ter)	SLC22A5	5	132378380	Renal carnitine transport defect
NM_003060.3(SLC22A5):c.844C>T (p.Arg282Ter)	SLC22A5	5	132387044	Renal carnitine transport defect
NM_003060.3(SLC22A5):c.1433C>T (p.Pro478Leu)	SLC22A5	5	132392598	Renal carnitine transport defect
NM_003060.3(SLC22A5):c.505C>T (p.Arg169Trp)	SLC22A5	5	132384154	Renal carnitine transport defect
NM_003060.3(SLC22A5):c.1196G>A (p.Arg399Gln)	SLC22A5	5	132390833	Renal carnitine transport defect
NM_003060.3(SLC22A5):c.760C>T (p.Arg254Ter)	SLC22A5	5	132385435	Renal carnitine transport defect
NM_003060.3(SLC22A5):c.1319C>T (p.Thr440Met)	SLC22A5	5	132392484	Renal carnitine transport defect
NM_003060.3(SLC22A5):c.506G>A (p.Arg169Gln)	SLC22A5	5	132384155	Renal carnitine transport defect;not provided
NM_003060.3(SLC22A5):c.1195C>T (p.Arg399Trp)	SLC22A5	5	132390832	Renal carnitine transport defect;not provided
NM_003060.3(SLC22A5):c.695C>T (p.Thr232Met)	SLC22A5	5	132385370	Renal carnitine transport defect;not provided
NM_003060.3(SLC22A5):c.845G>A (p.Arg282Gln)	SLC22A5	5	132387045	Renal carnitine transport defect;not provided
NM_003060.3(SLC22A5):c.1193C>T (p.Pro398Leu)	SLC22A5	5	132390830	Renal carnitine transport defect;not provided
NM_003060.3(SLC22A5):c.1463G>A (p.Arg488His)	SLC22A5	5	132393688	Renal carnitine transport defect;not provided
NM_003060.3(SLC22A5):c.338G>A (p.Cys113Tyr)	SLC22A5	5	132370310	Renal carnitine transport defect;not provided
NM_003060.3(SLC22A5):c.136C>T (p.Pro46Ser)	SLC22A5	5	132370108	Renal carnitine transport defect;not provided
NM_003990.4(PAX2):c.226G>A (p.Gly76Ser)	PAX2	10	100750707	Renal coloboma syndrome
NM_000537.3(REN):c.145C>T (p.Arg49Ter)	REN	1	204162117	Renal dysplasia
NM_000537.3(REN):c.689G>A (p.Arg230Lys)	REN	1	204159399	Renal dysplasia
NM_031850.3(AGTR1):c.950C>T (p.Thr317Met)	AGTR1	3	148741880	Renal dysplasia
NM_000029.3(AGT):c.1124G>A (p.Arg375Gln)	AGT	1	230705933	Renal dysplasia
NM_000029.3(AGT):c.604C>T (p.Gln202Ter)	AGT	1	230710247	Renal dysplasia
NM_031850.3(AGTR1):c.356G>A (p.Trp119Ter)	AGTR1	3	148741286	Renal dysplasia
NM_031850.3(AGTR1):c.481C>T (p.Arg161Ter)	AGTR1	3	148741411	Renal dysplasia
NM_000789.3(ACE):c.2371C>T (p.Arg791Ter)	ACE	17	63488713	Renal dysplasia
NM_000537.3(REN):c.127C>T (p.Arg43Ter)	REN	1	204162135	Renal dysplasia
NM_001080512.2(BICC1):c.259C>T (p.Gln87Ter)	BICC1	10	58702095	Renal dysplasia diffuse cystic
NM_014714.3(IFT140):c.1990G>A (p.Glu664Lys)	IFT140	16	1564074	Renal dysplasia, retinal pigmentary dystrophy, cerebellar ataxia and skeletal dysplasia
NM_014714.3(IFT140):c.874G>A (p.Val292Met)	IFT140	16	1587961	Renal dysplasia, retinal pigmentary dystrophy, cerebellar ataxia and skeletal dysplasia
NM_014714.3(IFT140):c.3991C>T (p.Gln1331Ter)	IFT140	16	1519930	Renal dysplasia, retinal pigmentary dystrophy, cerebellar ataxia and skeletal dysplasia
NM_014714.3(IFT140):c.634G>A (p.Gly212Arg)	IFT140	16	1592176	Renal dysplasia, retinal pigmentary dystrophy, cerebellar ataxia and skeletal dysplasia;not provided
NM_000789.3(ACE):c.1486C>T (p.Arg496Ter)	ACE	17	63483172	Renal dysplasia;not provided

NM_000789.3(ACE):c.1522C>T (p.Arg508Ter)	ACE	17	63483494	Renal dysplasia;not provided
NM_152383.4(DIS3L2):c.1466G>A (p.Cys489Tyr)	DIS3L2	2	232263247	Renal hamartomas nephroblastomatosis and fetal gigantism
NM_020041.2(SLC2A9):c.1138C>T (p.Arg380Trp)	SLC2A9	4	9890687	Renal hypouricemia 2
NM_020041.2(SLC2A9):c.511C>T (p.Arg171Cys)	SLC2A9	4	9985693	Renal hypouricemia 2
NM_020041.2(SLC2A9):c.374C>T (p.Thr125Met)	SLC2A9	4	9996817	Renal hypouricemia 2
NM_001692.3(ATP6V1B1):c.785+1G>A	ATP6V1B1	2	70961694	Renal tubular acidosis with progressive nerve deafness
NM_000342.3(SLC4A1):c.1766G>A (p.Arg589His)	SLC4A1	17	44255707	Renal tubular acidosis, distal, autosomal dominant
NM_000342.3(SLC4A1):c.1765C>T (p.Arg589Cys)	SLC4A1	17	44255708	Renal tubular acidosis, distal, autosomal dominant
NM_000342.3(SLC4A1):c.1838C>T (p.Ser613Phe)	SLC4A1	17	44255259	Renal tubular acidosis, distal, autosomal dominant
NM_020632.2(ATP6V0A4):c.2257C>T (p.Gln753Ter)	ATP6V0A4	7	138715764	Renal tubular acidosis, distal, autosomal recessive
NM_020632.2(ATP6V0A4):c.2458G>A (p.Gly820Arg)	ATP6V0A4	7	138706689	Renal tubular acidosis, distal, autosomal recessive
NM_020632.2(ATP6V0A4):c.1691+1G>A	ATP6V0A4	7	138734135	Renal tubular acidosis, distal, autosomal recessive
NM_020632.2(ATP6V0A4):c.418-1G>A	ATP6V0A4	7	138762435	Renal tubular acidosis, distal, autosomal recessive
NM_020632.2(ATP6V0A4):c.1571C>T (p.Pro524Leu)	ATP6V0A4	7	138739541	Renal tubular acidosis, distal, autosomal recessive
NM_020632.2(ATP6V0A4):c.2420G>A (p.Arg807Gln)	ATP6V0A4	7	138709633	Renal tubular acidosis, distal, autosomal recessive, with late-onset sensorineural hearing loss
NM_000342.3(SLC4A1):c.2102G>A (p.Gly701Asp)	SLC4A1	17	44253327	Renal tubular acidosis, distal, with hemolytic anemia
NM_000342.3(SLC4A1):c.1462G>A (p.Val488Met)	SLC4A1	17	44257514	Renal tubular acidosis, distal, with hemolytic anemia;Spherocytosis type 4
NM_001692.3(ATP6V1B1):c.91C>T (p.Arg31Ter)	ATP6V1B1	2	70936045	Renal tubular acidosis, distal, with progressive deafness
NM_001692.3(ATP6V1B1):c.232G>A (p.Gly78Arg)	ATP6V1B1	2	70958103	Renal tubular acidosis, distal, with progressive deafness
NM_001098484.2(SLC4A4):c.1661G>A (p.Arg554His)	SLC4A4	4	71472728	Renal tubular acidosis, proximal, with ocular abnormalities and mental retardation
NM_003759.3(SLC4A4):c.85C>T (p.Gln29Ter)	SLC4A4	4	71339333	Renal tubular acidosis, proximal, with ocular abnormalities and mental retardation
NM_153240.4(NPHP3):c.2918G>A (p.Arg973Gln)	NPHP3	3	132688857	Renal-hepatic-pancreatic dysplasia
NM_153240.4(NPHP3):c.3340C>T (p.Gln1114Ter)	NPHP3	3	132684784	Renal-hepatic-pancreatic dysplasia
NM_178170.2(NEK8):c.1795C>T (p.Arg599Ter)	NEK8	17	28741140	Renal-hepatic-pancreatic dysplasia 2
NM_013411.4(AK2):c.556C>T (p.Arg186Cys)	AK2	1	33013345	Reticular dysgenesis
NM_013411.4(AK2):c.307C>T (p.Arg103Trp)	AK2	1	33021616	Reticular dysgenesis
NM_001110.3(ADAM10):c.415C>T (p.Pro139Ser)	ADAM10	15	58679193	Reticulate acropigmentation of Kitamura
NM_001110.3(ADAM10):c.1511G>A (p.Ser504Asn)	ADAM10	15	58621471	Reticulate acropigmentation of Kitamura
NM_001110.3(ADAM10):c.1571G>A (p.Cys524Tyr)	ADAM10	15	58611932	Reticulate acropigmentation of Kitamura
NM_001553.2(IGFBP7):c.830-1G>A	IGFBP7	4	57031337	Retinal arterial macroaneurysm with supravascular pulmonic stenosis
NM_133497.3(KCNV2):c.1376G>A (p.Gly459Asp)	KCNV2	9	2729465	Retinal cone dystrophy 3B
NM_133497.3(KCNV2):c.226C>T (p.Gln76Ter)	KCNV2	9	2717965	Retinal cone dystrophy 3B
NR_029621.1(MIR204):n.37C>T	MIR204	9	70810048	Retinal dystrophy and iris coloboma with or without congenital cataract
NM_018191.3(RCBTB1):c.919G>A (p.Val307Met)	RCBTB1	13	49549584	RETINAL DYSTROPHY WITH OR WITHOUT EXTRAOCULAR ANOMALIES;Retinitis pigmentosa
NM_000350.2(ABCA4):c.6088C>T (p.Arg2030Ter)	ABCA4	1	94005500	Retinal dystrophy, early-onset severe;not provided
NM_006744.3(RBP4):c.278G>A (p.Gly93Asp)	RBP4	10	93600470	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome
NM_006744.3(RBP4):c.111+1G>A	RBP4	10	93600917	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome
NM_016026.3(RDH11):c.199C>T (p.Arg67Ter)	RDH11	14	67692588	Retinal dystrophy, juvenile cataracts, and short stature syndrome
NM_016026.3(RDH11):c.322C>T (p.Arg108Ter)	RDH11	14	67692465	Retinal dystrophy, juvenile cataracts, and short stature syndrome
NM_006017.2(PROM1):c.1726C>T (p.Gln576Ter)	PROM1	4	15994028	Retinal dystrophy;Retinitis pigmentosa 41
NM_000350.2(ABCA4):c.4469G>A (p.Cys1490Tyr)	ABCA4	1	94029515	Retinal dystrophy;Stargardt disease 1;not provided
NM_000322.4(PRPH2):c.410G>A (p.Gly137Asp)	PRPH2	6	42721925	Retinitis pigmentosa
NM_000539.3(RHO):c.520G>A (p.Gly174Ser)	RHO	3	129531034	Retinitis pigmentosa
NM_000539.3(RHO):c.562G>A (p.Gly188Arg)	RHO	3	129532282	Retinitis pigmentosa
NM_001142800.1(EYS):c.6557G>A (p.Gly2186Glu)	EYS	6	64081870	Retinitis pigmentosa

NM_001142800.1(EYS):c.7793G>A (p.Gly2598Asp)	EYS	6	63778111	Retinitis pigmentosa	
NM_001142800.1(EYS):c.7919G>A (p.Trp2640Ter)	EYS	6	63762613	Retinitis pigmentosa	
NM_004183.3(BEST1):c.763C>T (p.Arg255Trp)	BEST1	11	61958194	Retinitis pigmentosa	
NM_014014.4(SNRNP200):c.2042G>A (p.Arg681His)	SNRNP200	2	96293090	Retinitis pigmentosa	
NM_016247.3(IMPG2):c.3262C>T (p.Arg1088Ter)	IMPG2	3	101231117	Retinitis pigmentosa	
NM_001257965.1(CRB1):c.2711G>A (p.Cys904Tyr)	CRB1	1	197429555	Retinitis pigmentosa	
NM_014714.3(IFT140):c.3827G>A (p.Gly1276Glu)	IFT140	16	1520177	Retinitis pigmentosa	
NM_018191.3(RCBTB1):c.973C>T (p.His325Tyr)	RCBTB1	13	49549530	Retinitis pigmentosa	
NM_006269.1(RP1):c.2035C>T (p.Gln679Ter)	RP1	8	54625917	Retinitis pigmentosa 1	
NM_006269.1(RP1):c.2029C>T (p.Arg677Ter)	RP1	8	54625911	Retinitis pigmentosa 1;not provided	
NM_000883.3(IMPDH1):c.1057G>A (p.Val353Ile)	IMPDH1	7	128398431	Retinitis pigmentosa 10	
NM_000883.3(IMPDH1):c.931G>A (p.Asp311Asn)	IMPDH1	7	128398557	Retinitis pigmentosa 10;not provided	
NM_015629.3(PRPF31):c.1273C>T (p.Gln425Ter)	PRPF31	19	54129183	Retinitis pigmentosa 11	
NM_015629.3(PRPF31):c.1073+1G>A	PRPF31	19	54128201	Retinitis pigmentosa 11	
NM_201253.2(CRB1):c.2290C>T (p.Arg764Cys)	CRB1	1	197427615	Retinitis pigmentosa 12;not provided	
NM_201253.2(CRB1):c.2234C>T (p.Thr745Met)	CRB1	1	197427559	Retinitis pigmentosa 12;not provided	
NM_006445.3(PRPF8):c.6929G>A (p.Arg2310Lys)	PRPF8	17	1650881	Retinitis pigmentosa 13	
NM_006445.3(PRPF8):c.6353C>T (p.Ser2118Phe)	PRPF8	17	1653558	Retinitis pigmentosa 13	
NM_003322.5(TULP1):c.1444C>T (p.Arg482Trp)	TULP1	6	35500032	Retinitis pigmentosa 14	
NM_000328.2(RPGR):c.1387C>T (p.Gln463Ter)	RPGR	X	38297311	Retinitis pigmentosa 15	
NM_000328.2(RPGR):c.703C>T (p.Pro235Ser)	RPGR	X	38310690	Retinitis pigmentosa 15;not provided	
NM_000328.2(RPGR):c.823G>A (p.Gly275Ser)	RPGR	X	38304746	Retinitis pigmentosa 15;not provided	
NM_000717.4(CA4):c.206G>A (p.Arg69His)	CA4	17	60156653	Retinitis pigmentosa 17	
NM_004698.3(PRPF3):c.1481C>T (p.Thr494Met)	PRPF3	1	150344216	Retinitis pigmentosa 18	
NM_004698.3(PRPF3):c.1477C>T (p.Pro493Ser)	PRPF3	1	150344212	Retinitis pigmentosa 18	
NM_000350.2(ABCA4):c.1938-1G>A	ABCA4	1	94060760	Retinitis pigmentosa 19;Stargardt disease 1;not provided	
NM_000350.2(ABCA4):c.4577C>T (p.Thr1526Met)	ABCA4	1	94025011	Retinitis pigmentosa 19;Stargardt disease 1;not provided	
NM_000350.2(ABCA4):c.6229C>T (p.Arg2077Trp)	ABCA4	1	94001911	Retinitis pigmentosa 19;Stargardt disease 1;not provided	
NM_006915.2(RP2):c.76C>T (p.Gln26Ter)	RP2	X	46837176	Retinitis pigmentosa 2	
NM_006915.2(RP2):c.353G>A (p.Arg118His)	RP2	X	46853726	Retinitis pigmentosa 2	
NM_006915.2(RP2):c.358C>T (p.Arg120Ter)	RP2	X	46853731	Retinitis pigmentosa 2	
NM_000329.2(RPE65):c.271C>T (p.Arg91Trp)	RPE65	1	68444858	Retinitis pigmentosa 20;not provided	
NM_001142800.1(EYS):c.2194C>T (p.Gln732Ter)	EYS	6	64997647	Retinitis pigmentosa 25	
NM_001142800.1(EYS):c.490C>T (p.Arg164Ter)	EYS	6	65494921	Retinitis pigmentosa 25	
NM_006177.3(NRL):c.151C>T (p.Pro51Ser)	NRL	14	24082698	Retinitis pigmentosa 27	
NM_001201543.1(FAM161A):c.685C>T (p.Arg229Ter)	FAM161A	2	61840319	Retinitis pigmentosa 28	
NM_001201543.1(FAM161A):c.1786C>T (p.Arg596Ter)	FAM161A	2	61836075	Retinitis pigmentosa 28	
NM_001201543.1(FAM161A):c.1567C>T (p.Arg523Ter)	FAM161A	2	61839437	Retinitis pigmentosa 28;not provided	
NM_014014.4(SNRNP200):c.3260C>T (p.Ser1087Leu)	SNRNP200	2	96287968	Retinitis pigmentosa 33	
NM_001077620.2(PRCD):c.5G>A (p.Cys2Tyr)	PRCD	17	76540146	Retinitis pigmentosa 36	
NM_001077620.2(PRCD):c.64C>T (p.Arg22Ter)	PRCD	17	76540205	Retinitis pigmentosa 36	
NM_014249.3(NR2E3):c.166G>A (p.Gly56Arg)	NR2E3	15	71811530	Retinitis pigmentosa 37;not provided	
NM_006343.2(MERTK):c.1951C>T (p.Arg651Ter)	MERTK	2	112008466	Retinitis pigmentosa 38	
NM_006343.2(MERTK):c.2323C>T (p.Arg775Ter)	MERTK	2	112021555	Retinitis pigmentosa 38	
NM_206933.2(USH2A):c.2209C>T (p.Arg737Ter)	USH2A	1	216247185	Retinitis pigmentosa 39;Usher syndrome, type 2A	

NM_206933.2(USH2A):c.14803C>T (p.Arg4935Ter)	USH2A	1	215640723	Retinitis pigmentosa 39;Usher syndrome, type 2A
NM_206933.2(USH2A):c.10073G>A (p.Cys3358Tyr)	USH2A	1	215790168	Retinitis pigmentosa 39;Usher syndrome, type 2A;not provided
NM_000539.3(RHO):c.1040C>T (p.Pro347Leu)	RHO	3	129533711	Retinitis pigmentosa 4
NM_000539.3(RHO):c.1039C>T (p.Pro347Ser)	RHO	3	129533710	Retinitis pigmentosa 4
NM_000539.3(RHO):c.50C>T (p.Thr17Met)	RHO	3	129528783	Retinitis pigmentosa 4
NM_000539.3(RHO):c.266G>A (p.Gly89Asp)	RHO	3	129528999	Retinitis pigmentosa 4
NM_000539.3(RHO):c.568G>A (p.Asp190Asn)	RHO	3	129532288	Retinitis pigmentosa 4
NM_000539.3(RHO):c.1030C>T (p.Gln344Ter)	RHO	3	129533701	Retinitis pigmentosa 4
NM_000539.3(RHO):c.544G>A (p.Gly182Ser)	RHO	3	129532264	Retinitis pigmentosa 4
NM_000539.3(RHO):c.800C>T (p.Pro267Leu)	RHO	3	129532636	Retinitis pigmentosa 4
NM_000539.3(RHO):c.329G>A (p.Cys110Tyr)	RHO	3	129529062	Retinitis pigmentosa 4
NM_000539.3(RHO):c.316G>A (p.Gly106Arg)	RHO	3	129529049	Retinitis pigmentosa 4
NM_000539.3(RHO):c.341G>A (p.Gly114Asp)	RHO	3	129529074	Retinitis pigmentosa 4
NM_000539.3(RHO):c.511C>T (p.Pro171Ser)	RHO	3	129531025	Retinitis pigmentosa 4
NM_000539.3(RHO):c.1033G>A (p.Val345Met)	RHO	3	129533704	Retinitis pigmentosa 4
NM_000539.3(RHO):c.541G>A (p.Glu181Lys)	RHO	3	129532261	Retinitis pigmentosa 4
NM_000539.3(RHO):c.448G>A (p.Glu150Lys)	RHO	3	129530962	Retinitis pigmentosa 4, autosomal recessive
NM_000539.3(RHO):c.482G>A (p.Trp161Ter)	RHO	3	129530996	Retinitis pigmentosa 4, autosomal recessive
NM_000283.3(PDE6B):c.892C>T (p.Gln298Ter)	PDE6B	4	654119	Retinitis pigmentosa 40
NM_000283.3(PDE6B):c.1591C>T (p.Arg531Ter)	PDE6B	4	660590	Retinitis pigmentosa 40
NM_000283.3(PDE6B):c.1678C>T (p.Arg560Cys)	PDE6B	4	662197	Retinitis pigmentosa 40
NM_001031710.2(KLHL7):c.449G>A (p.Ser150Asn)	KLHL7	7	23140775	Retinitis pigmentosa 42
NM_001031710.2(KLHL7):c.457G>A (p.Ala153Thr)	KLHL7	7	23140783	Retinitis pigmentosa 42
NM_001031710.2(KLHL7):c.458C>T (p.Ala153Val)	KLHL7	7	23140784	Retinitis pigmentosa 42;not provided
NM_000440.2(PDE6A):c.1683G>A (p.Trp561Ter)	PDE6A	5	149895228	Retinitis pigmentosa 43
NM_000440.2(PDE6A):c.2053G>A (p.Val685Met)	PDE6A	5	149883511	Retinitis pigmentosa 43
NM_000440.2(PDE6A):c.1926+1G>A	PDE6A	5	149884779	Retinitis pigmentosa 43
NM_001297.4(CNGB1):c.2128C>T (p.Gln710Ter)	CNGB1	16	57917306	Retinitis pigmentosa 45
NM_001297.4(CNGB1):c.952C>T (p.Gln318Ter)	CNGB1	16	57950463	Retinitis pigmentosa 45;not provided
NM_000087.3(CNGA1):c.959C>T (p.Ser320Phe)	CNGA1	4	47937535	Retinitis pigmentosa 49
NM_004183.3(BEST1):c.682G>A (p.Asp228Asn)	BEST1	11	61957432	Retinitis pigmentosa 50;Retinitis pigmentosa, concentric;not provided
NM_001029883.2(C2orf71):c.556C>T (p.Gln186Ter)	C2orf71	2	29073706	Retinitis pigmentosa 54
NM_001029883.2(C2orf71):c.1828C>T (p.Gln610Ter)	C2orf71	2	29072434	Retinitis pigmentosa 54
NM_032146.5(ARL6):c.266C>T (p.Ala89Val)	ARL6	3	97784966	Retinitis pigmentosa 55
NM_016247.3(IMPG2):c.2716C>T (p.Arg906Ter)	IMPG2	3	101243615	Retinitis pigmentosa 56
NM_016247.3(IMPG2):c.2890C>T (p.Arg964Ter)	IMPG2	3	101242820	Retinitis pigmentosa 56
NM_012469.3(PRPF6):c.2185C>T (p.Arg729Trp)	PRPF6	20	64027138	Retinitis pigmentosa 60
NM_001195794.1(CLRN1):c.92C>T (p.Pro31Leu)	CLRN1	3	150972617	Retinitis pigmentosa 61
NM_001242957.2(MAK):c.37G>A (p.Gly13Ser)	MAK	6	10830612	Retinitis pigmentosa 62
NM_001242957.2(MAK):c.497G>A (p.Arg166His)	MAK	6	10803886	Retinitis pigmentosa 62
NM_177965.3(C8orf37):c.555G>A (p.Trp185Ter)	C8orf37	8	95247686	Retinitis pigmentosa 64
NM_002900.2(RBP3):c.3238G>A (p.Asp1080Asn)	RBP3	10	47353508	Retinitis pigmentosa 66
NM_020949.2(SLC7A14):c.988G>A (p.Gly330Arg)	SLC7A14	3	170483441	Retinitis pigmentosa 68
NM_020949.2(SLC7A14):c.395C>T (p.Ala132Val)	SLC7A14	3	170501255	Retinitis pigmentosa 68
NM_018474.4(KIZ):c.226C>T (p.Arg76Ter)	KIZ	20	21136463	Retinitis pigmentosa 69

NM_000322.4(PRPH2):c.647C>T (p.Pro216Leu)	PRPH2	6	42704546	Retinitis pigmentosa 7;not provided
NM_000322.4(PRPH2):c.136C>T (p.Arg46Ter)	PRPH2	6	42722199	Retinitis pigmentosa 7;not provided
NM_004697.4(PRPF4):c.944C>T (p.Pro315Leu)	PRPF4	9	113288183	Retinitis pigmentosa 70
NM_015662.2(IFT172):c.1525-1G>A	IFT172	2	27471096	Retinitis pigmentosa 71
NM_024741.2(ZNF408):c.1621C>T (p.Arg541Cys)	ZNF408	11	46705321	Retinitis pigmentosa 72
NM_021831.5(AGBL5):c.883G>A (p.Asp295Asn)	AGBL5	2	27055228	Retinitis pigmentosa 75
NM_021831.5(AGBL5):c.826C>T (p.Arg276Trp)	AGBL5	2	27055171	Retinitis pigmentosa 75
NM_001243766.1(POMGNT1):c.187C>T (p.Arg63Ter)	POMGNT1	1	46197018	RETINITIS PIGMENTOSA 76
NM_017739.3(POMGNT1):c.466G>A (p.Glu156Lys)	POMGNT1	1	46195879	RETINITIS PIGMENTOSA 76
NM_138393.2(REEP6):c.383C>T (p.Pro128Leu)	REEP6	19	1496319	RETINITIS PIGMENTOSA 77
NM_033500.2(HK1):c.2503G>A (p.Glu835Lys)	HK1	10	69398758	RETINITIS PIGMENTOSA 79
NM_000717.4(CA4):c.40C>T (p.Arg14Trp)	CA4	17	60150074	Retinitis Pigmentosa, Dominant;Retinitis pigmentosa 17
NM_201548.4(CERKL):c.769C>T (p.Arg257Ter)	CERKL	2	181558617	Retinitis Pigmentosa, Recessive;Retinitis pigmentosa 26
NM_000329.2(RPE65):c.118G>A (p.Gly40Ser)	RPE65	1	68446837	Retinitis pigmentosa;not provided
NM_000322.4(PRPH2):c.499G>A (p.Gly167Ser)	PRPH2	6	42721836	Retinitis pigmentosa;not provided
NM_000539.3(RHO):c.403C>T (p.Arg135Trp)	RHO	3	129530917	Retinitis pigmentosa;Retinitis pigmentosa 4;Retinitis punctata albescens;not provided
NM_000283.3(PDE6B):c.1669C>T (p.His557Tyr)	PDE6B	4	662188	Retinitis pigmentosa;Retinitis pigmentosa 40
NM_000283.3(PDE6B):c.2193+1G>A	PDE6B	4	664945	Retinitis pigmentosa;Retinitis pigmentosa 40
NM_152443.2(RDH12):c.377C>T (p.Ala126Val)	RDH12	14	67726084	Retinitis pigmentosa;Retinitis pigmentosa 53
NM_032119.3(ADGRV1):c.6901C>T (p.Gln2301Ter)	ADGRV1	5	90690991	Retinitis pigmentosa-deafness syndrome;Usher syndrome, type 2C
NM_000321.2(RB1):c.1333C>T (p.Arg445Ter)	RB1	13	48379594	Retinoblastoma
NM_000321.2(RB1):c.1700C>T (p.Ser567Leu)	RB1	13	48452997	Retinoblastoma
NM_000321.2(RB1):c.1072C>T (p.Arg358Ter)	RB1	13	48368549	Retinoblastoma
NM_000321.2(RB1):c.2212-1G>A	RB1	13	48464997	Retinoblastoma
NM_000321.2(RB1):c.-198G>A	RB1	13	48303715	Retinoblastoma
NM_000321.2(RB1):c.2211G>A (p.Glu737=)	RB1	13	48463835	Retinoblastoma
NM_000321.2(RB1):c.2101G>A (p.Asp701Asn)	RB1	13	48459828	Retinoblastoma
NM_000321.2(RB1):c.2197C>T (p.His733Tyr)	RB1	13	48463821	Retinoblastoma
NM_000321.2(RB1):c.1723C>T (p.Gln575Ter)	RB1	13	48453020	Retinoblastoma
NM_000321.2(RB1):c.1735C>T (p.Arg579Ter)	RB1	13	48453032	Retinoblastoma
NM_000321.2(RB1):c.1909C>T (p.Gln637Ter)	RB1	13	48456298	Retinoblastoma
NM_000321.2(RB1):c.1960+5G>A	RB1	13	48456354	Retinoblastoma
NM_000321.2(RB1):c.2490-1G>A	RB1	13	48473359	Retinoblastoma
NM_000321.2(RB1):c.2520+1G>A	RB1	13	48473391	Retinoblastoma
NM_000321.2(RB1):c.103C>T (p.Gln35Ter)	RB1	13	48304015	Retinoblastoma
NM_000321.2(RB1):c.958C>T (p.Arg320Ter)	RB1	13	48367512	Retinoblastoma
NM_000321.2(RB1):c.1049+1G>A	RB1	13	48367604	Retinoblastoma
NM_000321.2(RB1):c.1215+1G>A	RB1	13	48373493	Retinoblastoma
NM_000321.2(RB1):c.1216-1G>A	RB1	13	48376917	Retinoblastoma
NM_000321.2(RB1):c.1332+1G>A	RB1	13	48377035	Retinoblastoma
NM_000321.2(RB1):c.1654C>T (p.Arg552Ter)	RB1	13	48381402	Retinoblastoma
NM_000321.2(RB1):c.1688G>A (p.Trp563Ter)	RB1	13	48381436	Retinoblastoma
NM_000321.2(RB1):c.1695+1G>A	RB1	13	48381444	Retinoblastoma
NM_000321.2(RB1):c.1696-1G>A	RB1	13	48452992	Retinoblastoma
NM_000321.2(RB1):c.297G>A (p.Trp99Ter)	RB1	13	48342631	Retinoblastoma

NM_000321.2(RB1):c.2053C>T (p.Gln685Ter)	RB1	13	48459780	Retinoblastoma
NM_000321.2(RB1):c.2548C>T (p.Gln850Ter)	RB1	13	48476728	Retinoblastoma
NM_000321.2(RB1):c.1510C>T (p.Gln504Ter)	RB1	13	48381258	Retinoblastoma
NM_000321.2(RB1):c.2359C>T (p.Arg787Ter)	RB1	13	48465238	Retinoblastoma;not provided
NM_000321.2(RB1):c.1981C>T (p.Arg661Trp)	RB1	13	48459708	Retinoblastoma;not provided
NM_000321.2(RB1):c.1399C>T (p.Arg467Ter)	RB1	13	48380062	Retinoblastoma;not provided
NM_000321.2(RB1):c.763C>T (p.Arg255Ter)	RB1	13	48362859	Retinoblastoma;not provided
NM_000321.2(RB1):c.1363C>T (p.Arg455Ter)	RB1	13	48379624	Retinoblastoma;not provided
NM_001110792.1(MECP2):c.5C>T (p.Ala2Val)	MECP2	X	154097661	Rett syndrome
NM_004992.3(MECP2):c.1357C>T (p.Arg453Ter)	MECP2	X	154030471	Rett syndrome
NM_004992.3(MECP2):c.302C>T (p.Pro101Leu)	MECP2	X	154032282	Rett syndrome
NM_004992.3(MECP2):c.311G>A (p.Trp104Ter)	MECP2	X	154032273	Rett syndrome
NM_004992.3(MECP2):c.317G>A (p.Arg106Gln)	MECP2	X	154032267	Rett syndrome
NM_004992.3(MECP2):c.382C>T (p.Gln128Ter)	MECP2	X	154031446	Rett syndrome
NM_004992.3(MECP2):c.508C>T (p.Gln170Ter)	MECP2	X	154031320	Rett syndrome
NM_004992.3(MECP2):c.55C>T (p.Gln19Ter)	MECP2	X	154032529	Rett syndrome
NM_004992.3(MECP2):c.622C>T (p.Gln208Ter)	MECP2	X	154031206	Rett syndrome
NM_004992.3(MECP2):c.784C>T (p.Gln262Ter)	MECP2	X	154031044	Rett syndrome
NM_004992.3(MECP2):c.889C>T (p.Gln297Ter)	MECP2	X	154030939	Rett syndrome
NM_001110792.1(MECP2):c.48C>T (p.Gly16=)	MECP2	X	154097618	Rett syndrome
NM_005249.4(FOXG1):c.765G>A (p.Trp255Ter)	FOXG1	14	28768044	Rett syndrome, congenital variant
NM_005249.4(FOXG1):c.924G>A (p.Trp308Ter)	FOXG1	14	28768203	Rett syndrome, congenital variant
NM_005249.4(FOXG1):c.577G>A (p.Ala193Thr)	FOXG1	14	28767856	Rett syndrome, congenital variant
NM_005249.4(FOXG1):c.689G>A (p.Arg230His)	FOXG1	14	28767968	Rett syndrome, congenital variant
NM_005249.4(FOXG1):c.730C>T (p.Arg244Cys)	FOXG1	14	28768009	Rett syndrome, congenital variant
NM_005249.4(FOXG1):c.821G>A (p.Arg274Gln)	FOXG1	14	28768100	Rett syndrome, congenital variant
NM_005249.4(FOXG1):c.256C>T (p.Gln86Ter)	FOXG1	14	28767535	Rett syndrome, congenital variant;not provided
NM_004992.3(MECP2):c.316C>T (p.Arg106Trp)	MECP2	X	154032268	Rett syndrome;not provided
NM_004992.3(MECP2):c.917G>A (p.Arg306His)	MECP2	X	154030911	Rett syndrome;not provided
NM_004992.3(MECP2):c.377+1G>A	MECP2	X	154032206	Rett syndrome;not provided
NM_194442.2(LBR):c.1114C>T (p.Arg372Cys)	LBR	1	225411411	Reynolds syndrome
NM_003073.4(SMARCB1):c.*82C>T	SMARCB1	22	23834262	Rhabdoid tumor predisposition syndrome 1
NM_003073.4(SMARCB1):c.118C>T (p.Arg40Ter)	SMARCB1	22	23791780	Rhabdoid tumor predisposition syndrome 1;Schwannomatosis
NM_003073.4(SMARCB1):c.152G>A (p.Trp51Ter)	SMARCB1	22	23791814	Rhabdoid tumor predisposition syndrome 1;Schwannomatosis
NM_001128844.1(SMARCA4):c.3565C>T (p.Arg1189Ter)	SMARCA4	19	11033308	Rhabdoid tumor predisposition syndrome 2
NM_003072.3(SMARCA4):c.3533G>A (p.Trp1178Ter)	SMARCA4	19	11030880	Rhabdoid tumor predisposition syndrome 2
NM_001128845.1(SMARCA4):c.4071+1G>A	SMARCA4	19	11035133	Rhabdoid tumor predisposition syndrome 2
NM_001128849.1(SMARCA4):c.643C>T (p.Gln215Ter)	SMARCA4	19	10986476	Rhabdoid tumor predisposition syndrome 2
NM_001128849.1(SMARCA4):c.3239G>A (p.Gly1080Asp)	SMARCA4	19	11027807	Rhabdoid tumor predisposition syndrome 2
NM_001128849.1(SMARCA4):c.4339C>T (p.Arg1447Ter)	SMARCA4	19	11041379	Rhabdoid tumor predisposition syndrome 2
NM_004304.4(ALK):c.3673G>A (p.Asp1225Asn)	ALK	2	29214054	Rhabdomyosarcoma
NM_002555.5(SLC22A18):c.257G>A (p.Arg86His)	SLC22A18	11	2909210	Rhabdomyosarcoma, somatic
NM_001844.4(COL2A1):c.952G>A (p.Gly318Arg)	COL2A1	12	47993475	Rhegmatogenous retinal detachment, autosomal dominant
NM_001844.4(COL2A1):c.1957C>T (p.Arg653Ter)	COL2A1	12	47983721	Rhegmatogenous retinal detachment, autosomal dominant;Stickler syndrome type 1;not provided

NM_000288.3(PEX7):c.694C>T (p.Arg232Ter)	PEX7	6	136869950	Rhizomelic chondrodysplasia punctata type 1;not provided
NM_014236.3(GNPAT):c.632G>A (p.Arg211His)	GNPAT	1	231265356	Rhizomelic chondrodysplasia punctata type 2
NM_014236.3(GNPAT):c.631C>T (p.Arg211Cys)	GNPAT	1	231265355	Rhizomelic chondrodysplasia punctata type 2
NM_003659.3(AGPS):c.1256G>A (p.Arg419His)	AGPS	2	177493170	Rhizomelic chondrodysplasia punctata type 3
NM_003659.3(AGPS):c.926C>T (p.Thr309Ile)	AGPS	2	177461948	Rhizomelic chondrodysplasia punctata type 3
NM_003659.3(AGPS):c.1703C>T (p.Thr568Met)	AGPS	2	177521274	Rhizomelic chondrodysplasia punctata type 3
NM_000324.2(RHAG):c.236G>A (p.Ser79Asn)	RHAG	6	49619284	Rh-null hemolytic anemia, regulator type
NM_000324.2(RHAG):c.836G>A (p.Gly279Glu)	RHAG	6	49612506	Rh-null hemolytic anemia, regulator type
NM_152617.3(RNF168):c.391C>T (p.Arg131Ter)	RNF168	3	196487566	Riddle syndrome
NM_152743.3(BRAT1):c.1857G>A (p.Trp619Ter)	BRAT1	7	2538678	Rigidity and multifocal seizure syndrome, lethal neonatal
NM_153427.2(PITX2):c.185G>A (p.Arg62His)	PITX2	4	110621231	Ring dermoid of cornea
NM_033337.2(CAV3):c.139G>A (p.Glu47Lys)	CAV3	3	8745550	Rippling muscle disease 2;not provided
NM_001017420.2(ESCO2):c.505C>T (p.Arg169Ter)	ESCO2	8	27776813	Roberts-SC phocomelia syndrome
NM_001017420.2(ESCO2):c.1269G>A (p.Trp423Ter)	ESCO2	8	27791968	Roberts-SC phocomelia syndrome
NM_001017420.2(ESCO2):c.604C>T (p.Gln202Ter)	ESCO2	8	27776912	Roberts-SC phocomelia syndrome
NM_001017420.2(ESCO2):c.1131+1G>A	ESCO2	8	27788003	Roberts-SC phocomelia syndrome
NM_001017420.2(ESCO2):c.1354-18G>A	ESCO2	8	27792650	Roberts-SC phocomelia syndrome
NM_000474.3(TWIST1):c.211C>T (p.Gln71Ter)	TWIST1	7	19117111	Robinow Sorauf syndrome
NM_003392.4(WNT5A):c.206G>A (p.Cys69Tyr)	WNT5A	3	55479499	Robinow syndrome
NM_004560.3(ROR2):c.1504C>T (p.Gln502Ter)	ROR2	9	91724990	Robinow syndrome, autosomal recessive
NM_004560.3(ROR2):c.550C>T (p.Arg184Cys)	ROR2	9	91737463	Robinow syndrome, autosomal recessive
NM_004560.3(ROR2):c.2160G>A (p.Trp720Ter)	ROR2	9	91724334	Robinow syndrome, autosomal recessive
NM_004560.3(ROR2):c.613C>T (p.Arg205Ter)	ROR2	9	91737400	Robinow syndrome, autosomal recessive
NM_004560.3(ROR2):c.1324C>T (p.Arg442Ter)	ROR2	9	91726603	Robinow syndrome, autosomal recessive, with brachy-syn-polydactyly
NM_004560.3(ROR2):c.355C>T (p.Arg119Ter)	ROR2	9	91757380	Robinow syndrome, autosomal recessive;not provided
NM_004423.3(DVL3):c.1715-1G>A	DVL3	3	184170318	Robinow syndrome;Robinow syndrome, autosomal dominant 3
NR_023343.1(RNU4ATAC):n.16G>A	RNU4ATAC	2	121530895	Roifman syndrome
NR_023343.1(RNU4ATAC):n.13C>T	RNU4ATAC	2	121530892	Roifman syndrome
NR_023343.1(RNU4ATAC):n.37G>A	RNU4ATAC	2	121530916	Roifman syndrome
NR_023343.1(RNU4ATAC):n.48G>A	RNU4ATAC	2	121530927	Roifman syndrome
NR_023343.1(RNU4ATAC):n.8C>T	RNU4ATAC	2	121530887	Roifman syndrome
NM_004260.3(RECQL4):c.1391-1G>A	RECQL4	8	144515243	Rothmund-Thomson syndrome
NM_004260.3(RECQL4):c.1704+1G>A	RECQL4	8	144514441	Rothmund-Thomson syndrome
NM_004260.3(RECQL4):c.82C>T (p.Gln28Ter)	RECQL4	8	144517703	Rothmund-Thomson syndrome
NM_004260.3(RECQL4):c.1259-1G>A	RECQL4	8	144515458	Rothmund-Thomson syndrome
NM_006446.4(SLCO1B1):c.1738C>T (p.Arg580Ter)	SLCO1B1	12	21222355	Rotor syndrome
NM_006446.4(SLCO1B1):c.757C>T (p.Arg253Ter)	SLCO1B1	12	21196975	Rotor syndrome
NM_015713.4(RRM2B):c.122G>A (p.Arg41Gln)	RRM2B	8	102232231	RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.48G>A (p.Glu16=)	RRM2B	8	102238827	RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.121C>T (p.Arg41Trp)	RRM2B	8	102232232	RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.208G>A (p.Asp70Asn)	RRM2B	8	102226031	RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.328C>T (p.Arg110Cys)	RRM2B	8	102225012	RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.391G>A (p.Glu131Lys)	RRM2B	8	102224949	RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.431C>T (p.Thr144Ile)	RRM2B	8	102224909	RRM2B-related mitochondrial disease
NM_015713.4(RRM2B):c.583G>A (p.Gly195Arg)	RRM2B	8	102218915	RRM2B-related mitochondrial disease

NM_015713.4(RRM2B):c.632G>A (p.Arg211Lys)	RRM2B	8	102218866	RRM2B-related mitochondrial disease
NM_004380.2(CREBBP):c.406C>T (p.Gln136Ter)	CREBBP	16	3850689	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.1069C>T (p.Gln357Ter)	CREBBP	16	3793533	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.6088C>T (p.Gln2030Ter)	CREBBP	16	3728959	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.5821C>T (p.Gln1941Ter)	CREBBP	16	3729226	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.4133+1G>A	CREBBP	16	3740398	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.4078C>T (p.Arg1360Ter)	CREBBP	16	3740454	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.4045C>T (p.Gln1349Ter)	CREBBP	16	3740487	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.3836+1G>A	CREBBP	16	3749626	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.3310C>T (p.Gln1104Ter)	CREBBP	16	3758913	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.2791C>T (p.Gln931Ter)	CREBBP	16	3770659	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.1270C>T (p.Arg424Ter)	CREBBP	16	3792041	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.1257G>A (p.Trp419Ter)	CREBBP	16	3792054	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.1156C>T (p.Arg386Ter)	CREBBP	16	3793446	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.1063C>T (p.Gln355Ter)	CREBBP	16	3793539	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.598C>T (p.Gln200Ter)	CREBBP	16	3850497	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.316C>T (p.Gln106Ter)	CREBBP	16	3850779	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.286C>T (p.Gln96Ter)	CREBBP	16	3850809	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.5027G>A (p.Trp1676Ter)	CREBBP	16	3731337	Rubinstein-Taybi syndrome
NM_004380.2(CREBBP):c.3436C>T (p.Gln1146Ter)	CREBBP	16	3757982	Rubinstein-Taybi syndrome
NM_001429.3(EP300):c.1942C>T (p.Arg648Ter)	EP300	22	41141111	Rubinstein-Taybi syndrome 2
NM_001429.3(EP300):c.4933C>T (p.Arg1645Ter)	EP300	22	41176400	Rubinstein-Taybi syndrome 2
NM_004380.2(CREBBP):c.3832G>A (p.Glu1278Lys)	CREBBP	16	3749631	Rubinstein-Taybi syndrome;not provided
NM_004380.2(CREBBP):c.3779+1G>A	CREBBP	16	3751725	Rubinstein-Taybi syndrome;not provided
NM_000474.3(TWIST1):c.82C>T (p.Gln28Ter)	TWIST1	7	19117240	Saethre-Chotzen syndrome with eyelid anomalies
NM_012434.4(SLC17A5):c.115C>T (p.Arg39Cys)	SLC17A5	6	73644583	Salla disease;not provided
NM_012434.4(SLC17A5):c.291G>A (p.Thr97=)	SLC17A5	6	73644407	Salla disease;not provided
NM_012434.4(SLC17A5):c.819+1G>A	SLC17A5	6	73635381	Salla disease;not provided
NM_000521.3(HEXB):c.508C>T (p.Arg170Ter)	HEXB	5	74693701	Sandhoff disease
NM_000521.3(HEXB):c.1514G>A (p.Arg505Gln)	HEXB	5	74720648	Sandhoff disease, adult type
NM_000521.3(HEXB):c.1510C>T (p.Pro504Ser)	HEXB	5	74720644	Sandhoff disease, chronic
NM_000521.3(HEXB):c.1250C>T (p.Pro417Leu)	HEXB	5	74718804	Sandhoff disease;Sandhoff disease, adult type;Sandhoff disease, juvenile type;not provided
NM_000521.3(HEXB):c.850C>T (p.Arg284Ter)	HEXB	5	74713584	Sandhoff disease;Sandhoff disease, infantile
NM_001258221.1(KCTD1):c.92C>T (p.Pro31Leu)	KCTD1	18	26501144	Scalp ear nipple syndrome
NM_001258221.1(KCTD1):c.185G>A (p.Gly62Asp)	KCTD1	18	26476639	Scalp ear nipple syndrome
NM_001258221.1(KCTD1):c.58C>T (p.Pro20Ser)	KCTD1	18	26501178	Scalp ear nipple syndrome
NM_019066.4(MAGEL2):c.3124C>T (p.Gln1042Ter)	MAGEL2	15	23644619	Schaaf-yang syndrome;not provided
NM_014140.3(SMARCAL1):c.49C>T (p.Arg17Ter)	SMARCAL1	2	216414753	Schimke immunoosseous dysplasia
NM_014140.3(SMARCAL1):c.100C>T (p.Gln34Ter)	SMARCAL1	2	216414804	Schimke immunoosseous dysplasia
NM_014140.3(SMARCAL1):c.1933C>T (p.Arg645Cys)	SMARCAL1	2	216450927	Schimke immunoosseous dysplasia
NM_014140.3(SMARCAL1):c.1756C>T (p.Arg586Trp)	SMARCAL1	2	216447063	Schimke immunoosseous dysplasia
NM_014140.3(SMARCAL1):c.2291G>A (p.Arg764Gln)	SMARCAL1	2	216475315	Schimke immunoosseous dysplasia
NM_000262.2(NAGA):c.973G>A (p.Glu325Lys)	NAGA	22	42061052	Schindler disease, type 1;not provided
NM_015559.2(SETBP1):c.2602G>A (p.Asp868Asn)	SETBP1	18	44951942	Schinzel-Giedion syndrome
NM_015559.2(SETBP1):c.2609G>A (p.Gly870Asp)	SETBP1	18	44951949	Schinzel-Giedion syndrome

NM_001845.5(COL4A1):c.3976G>A (p.Gly1326Arg)	COL4A1	13	110166277	SCHIZENCEPHALY
NM_014712.2(SETD1A):c.2209C>T (p.Gln737Ter)	SETD1A	16	30966090	Schizophrenia
NM_033517.1(SHANK3):c.3349C>T (p.Arg1117Ter)	SHANK3	22	50721182	Schizophrenia 15
NM_033517.1(SHANK3):c.1606C>T (p.Arg536Trp)	SHANK3	22	50698797	Schizophrenia 15
NM_015139.2(SLC35D1):c.932G>A (p.Trp311Ter)	SLC35D1	1	67009112	Schneckenbecken dysplasia
NM_015139.2(SLC35D1):c.319C>T (p.Arg107Ter)	SLC35D1	1	67052776	Schneckenbecken dysplasia
NM_013319.2(UBIAD1):c.524C>T (p.Thr175Ile)	UBIAD1	1	11274055	Schnyder crystalline corneal dystrophy
NM_013319.2(UBIAD1):c.556G>A (p.Gly186Arg)	UBIAD1	1	11285670	Schnyder crystalline corneal dystrophy
NM_013319.2(UBIAD1):c.530G>A (p.Gly177Glu)	UBIAD1	1	11285644	Schnyder crystalline corneal dystrophy
NM_003073.4(SMARCB1):c.34C>T (p.Gln12Ter)	SMARCB1	22	23787203	Schwannomatosis 1
NM_003073.4(SMARCB1):c.143C>T (p.Pro48Leu)	SMARCB1	22	23791805	Schwannomatosis 1
NM_003073.4(SMARCB1):c.544C>T (p.Gln182Ter)	SMARCB1	22	23803338	Schwannomatosis 1, somatic
NM_006767.3(LZTR1):c.264-13G>A	LZTR1	22	20985828	Schwannomatosis 2
NM_006767.3(LZTR1):c.365C>T (p.Ser122Leu)	LZTR1	22	20987548	Schwannomatosis 2
NM_006767.3(LZTR1):c.2062C>T (p.Arg688Cys)	LZTR1	22	20995865	Schwannomatosis 2
NM_006767.3(LZTR1):c.1397G>A (p.Arg466Gln)	LZTR1	22	20993967	Schwannomatosis 2
NM_006767.3(LZTR1):c.1084C>T (p.Arg362Ter)	LZTR1	22	20992304	Schwannomatosis 2
NM_005529.6(HSPG2):c.4595G>A (p.Cys1532Tyr)	HSPG2	1	21864874	Schwartz Jampel syndrome type 1
NM_000022.3(ADA):c.467G>A (p.Arg156His)	ADA	20	44625580	SCID due to ADA deficiency, delayed onset;Severe combined immunodeficiency due to ADA deficiency
NM_025237.2(SOST):c.70C>T (p.Gln24Ter)	SOST	17	43758672	Sclerosteosis
NM_025237.2(SOST):c.372G>A (p.Trp124Ter)	SOST	17	43755612	Sclerosteosis
NM_025237.2(SOST):c.376C>T (p.Arg126Ter)	SOST	17	43755608	Sclerosteosis
NM_002334.3(LRP4):c.3508C>T (p.Arg1170Trp)	LRP4	11	46876494	Sclerosteosis 2
NM_002894.2(RBBP8):c.298C>T (p.Arg100Trp)	RBBP8	18	22968855	Seckel syndrome 2
NM_018451.4(CENPJ):c.1969C>T (p.Gln657Ter)	CENPJ	13	24906069	Seckel syndrome 4
NM_025180.3(CEP63):c.1068-1G>A	CEP63	3	134549061	Seckel syndrome 6
NM_005879.2(TRAIP):c.553C>T (p.Arg185Ter)	TRAIP	3	49841890	Seckel syndrome 9
NM_005879.2(TRAIP):c.52C>T (p.Arg18Cys)	TRAIP	3	49856402	Seckel syndrome 9
NM_000549.4(TSHB):c.145G>A (p.Gly49Arg)	TSHB	1	115033507	Secondary hypothyroidism
NM_000549.4(TSHB):c.205C>T (p.Gln69Ter)	TSHB	1	115034015	Secondary hypothyroidism
NM_199292.2(TH):c.698G>A (p.Arg233His)	TH	11	2167905	Segawa syndrome, autosomal recessive
NM_199292.2(TH):c.1010G>A (p.Arg337His)	TH	11	2166693	Segawa syndrome, autosomal recessive
NM_199292.2(TH):c.941C>T (p.Thr314Met)	TH	11	2166762	Segawa syndrome, autosomal recessive
NM_000360.3(TH):c.601C>T (p.Gln201Ter)	TH	11	2167909	Segawa syndrome, autosomal recessive
NM_014191.3(SCN8A):c.4447G>A (p.Glu1483Lys)	SCN8A	12	51790425	Seizures, benign familial infantile, 5
NM_005219.4(DIAPH1):c.3145C>T (p.Arg1049Ter)	DIAPH1	5	141528456	Seizures, cortical blindness, and microcephaly syndrome;not provided
NM_021007.2(SCN2A):c.2567G>A (p.Arg856Gln)	SCN2A	2	165344559	Seizures;not provided
NM_002448.3(MSX1):c.577C>T (p.Gln193Ter)	MSX1	4	4862808	Selective tooth agenesis 1
NM_001023570.3(IQCB1):c.1069C>T (p.Gln357Ter)	IQCB1	3	121790133	Senior-Loken syndrome 5
NM_001023570.3(IQCB1):c.1465C>T (p.Arg489Ter)	IQCB1	3	121772659	Senior-Loken syndrome 5
NM_001023570.3(IQCB1):c.214C>T (p.Arg72Ter)	IQCB1	3	121828519	Senior-Loken syndrome 5
NM_001023570.3(IQCB1):c.1090C>T (p.Arg364Ter)	IQCB1	3	121790112	Senior-Loken syndrome 5;not provided
NM_006642.4(SDCCAG8):c.481C>T (p.Gln161Ter)	SDCCAG8	1	243286332	Senior-Loken syndrome 7
NM_006642.4(SDCCAG8):c.567G>A (p.Trp189Ter)	SDCCAG8	1	243293111	Senior-Loken syndrome 7
NM_025132.3(WDR19):c.3565+1G>A	WDR19	4	39273062	Senior-Loken syndrome 8

NM_025132.3(WDR19):c.3533G>A (p.Arg1178Gln)	WDR19	4	39273029	Senior-Loken syndrome 8;not provided
NM_015650.3(TRAF3IP1):c.463C>T (p.Arg155Ter)	TRAF3IP1	2	238328794	Senior-Loken syndrome 9
NM_015650.3(TRAF3IP1):c.373G>A (p.Val125Met)	TRAF3IP1	2	238328704	Senior-Loken syndrome 9
NM_002693.2(POLG):c.1879C>T (p.Arg627Trp)	POLG	15	89325520	Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis
NM_002693.2(POLG):c.2794C>T (p.His932Tyr)	POLG	15	89320953	Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis
NM_003124.4(SPR):c.355C>T (p.Gln119Ter)	SPR	2	72888364	Sepiapterin reductase deficiency
NM_003124.4(SPR):c.488C>T (p.Pro163Leu)	SPR	2	72888497	Sepiapterin reductase deficiency
NM_003865.2(HESX1):c.478C>T (p.Arg160Cys)	HESX1	3	57198277	Septo-optic dysplasia sequence
NM_003865.2(HESX1):c.509C>T (p.Ser170Leu)	HESX1	3	57198246	Septooptic dysplasia, mild
NM_030761.4(WNT4):c.341C>T (p.Ala114Val)	WNT4	1	22121549	Serkal syndrome
NM_002241.4(KCNJ10):c.595C>T (p.Arg199Ter)	KCNJ10	1	160041938	SeSAME syndrome
NM_002241.4(KCNJ10):c.491C>T (p.Thr164Ile)	KCNJ10	1	160042042	SeSAME syndrome
NM_002241.4(KCNJ10):c.500C>T (p.Ala167Val)	KCNJ10	1	160042033	SeSAME syndrome
NM_002241.4(KCNJ10):c.193C>T (p.Arg65Cys)	KCNJ10	1	160042340	SeSAME syndrome
NM_002241.4(KCNJ10):c.524G>A (p.Arg175Gln)	KCNJ10	1	160042009	SeSAME syndrome
NM_002241.4(KCNJ10):c.889C>T (p.Arg297Cys)	KCNJ10	1	160041644	SeSAME syndrome;not provided
NM_000231.2(SGCG):c.848G>A (p.Cys283Tyr)	SGCG	13	23324513	Severe autosomal recessive muscular dystrophy of childhood - North African type
NM_000231.2(SGCG):c.787G>A (p.Glu263Lys)	SGCG	13	23324452	Severe autosomal recessive muscular dystrophy of childhood - North African type;not provided
NM_002185.3(IL7R):c.353G>A (p.Cys118Tyr)	IL7R	5	35867437	Severe combined immunodeficiency disease
NM_000215.3(JAK3):c.1767C>T (p.Gly589=)	JAK3	19	17837148	Severe combined immunodeficiency disease;not provided
NM_000448.2(RAG1):c.322C>T (p.Arg108Ter)	RAG1	11	36573626	Severe combined immunodeficiency disease;Severe immunodeficiency, autosomal recessive, T-cell negative, B-cell negative, NK cell-positive
NM_000022.3(ADA):c.301C>T (p.Arg101Trp)	ADA	20	44626517	Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.302G>A (p.Arg101Gln)	ADA	20	44626516	Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.632G>A (p.Arg211His)	ADA	20	44623053	Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.466C>T (p.Arg156Cys)	ADA	20	44625581	Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.872C>T (p.Ser291Leu)	ADA	20	44621121	Severe combined immunodeficiency due to ADA deficiency
NM_000022.3(ADA):c.986C>T (p.Ala329Val)	ADA	20	44620391	Severe combined immunodeficiency due to ADA deficiency;not provided
NM_000022.3(ADA):c.646G>A (p.Gly216Arg)	ADA	20	44623039	Severe combined immunodeficiency due to ADA deficiency;not provided
NM_024782.2(NHEJ1):c.532C>T (p.Arg178Ter)	NHEJ1	2	219146736	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation;not provided
NM_001033855.2(DCLRE1C):c.194C>T (p.Thr65Ile)	DCLRE1C	10	14945157	Severe combined immunodeficiency with sensitivity to ionizing radiation
NM_001033855.2(DCLRE1C):c.241C>T (p.Arg81Ter)	DCLRE1C	10	14945110	Severe combined immunodeficiency with sensitivity to ionizing radiation;not provided
NM_001079.3(ZAP70):c.1624-11G>A	ZAP70	2	97737984	Severe combined immunodeficiency, atypical
NM_001079.3(ZAP70):c.1394G>A (p.Arg465His)	ZAP70	2	97737577	Severe combined immunodeficiency, atypical
NM_001079.3(ZAP70):c.1393C>T (p.Arg465Cys)	ZAP70	2	97737576	Severe combined immunodeficiency, atypical
NM_001079.3(ZAP70):c.837+121G>A	ZAP70	2	97733464	Severe combined immunodeficiency, atypical
NM_000215.3(JAK3):c.1333C>T (p.Arg445Ter)	JAK3	19	17839585	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-negative
NM_002185.3(IL7R):c.651G>A (p.Trp217Ter)	IL7R	5	35873593	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive
NM_002185.3(IL7R):c.394C>T (p.Pro132Ser)	IL7R	5	35871070	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive
NM_000536.3(RAG2):c.1433G>A (p.Cys478Tyr)	RAG2	11	36592736	Severe combined immunodeficiency, B cell-negative
NM_000448.2(RAG1):c.2164G>A (p.Glu722Lys)	RAG1	11	36575468	Severe combined immunodeficiency, B cell-negative

NM_000448.2(RAG1):c.2326C>T (p.Arg776Trp)	RAG1	11	36575630	Severe combined immunodeficiency, B cell-negative
NM_032492.3(JAGN1):c.40G>A (p.Gly14Ser)	JAGN1	3	9890762	Severe congenital neutropenia
NM_032492.3(JAGN1):c.59G>A (p.Arg20Gln)	JAGN1	3	9890781	Severe congenital neutropenia
NM_006118.3(HAX1):c.568C>T (p.Gln190Ter)	HAX1	1	154275165	Severe congenital neutropenia 3, autosomal recessive
NM_006118.3(HAX1):c.256C>T (p.Arg86Ter)	HAX1	1	154273538	Severe congenital neutropenia 3, autosomal recessive
NM_138387.3(G6PC3):c.758G>A (p.Arg253His)	G6PC3	17	44075760	Severe congenital neutropenia 4, autosomal recessive
NM_138387.3(G6PC3):c.130C>T (p.Pro44Ser)	G6PC3	17	44071095	Severe congenital neutropenia 4, autosomal recessive
NM_138387.3(G6PC3):c.829C>T (p.Gln277Ter)	G6PC3	17	44075831	Severe congenital neutropenia 4, autosomal recessive
NM_007259.4(VPS45):c.712G>A (p.Glu238Lys)	VPS45	1	150081366	Severe congenital neutropenia 5, autosomal recessive
NM_001972.3(ELANE):c.214G>A (p.Val72Met)	ELANE	19	853022	Severe congenital neutropenia autosomal dominant
NM_001972.3(ELANE):c.377C>T (p.Ser126Leu)	ELANE	19	855574	Severe congenital neutropenia autosomal dominant
NM_001972.3(ELANE):c.416C>T (p.Pro139Leu)	ELANE	19	855613	Severe congenital neutropenia autosomal dominant;not provided
NM_001972.3(ELANE):c.640G>A (p.Gly214Arg)	ELANE	19	856000	Severe congenital neutropenia autosomal dominant;not provided
NM_000377.2(WAS):c.777+1G>A	WAS	X	48688097	Severe congenital neutropenia X-linked;Thrombocytopenia, X-linked;Wiskott-Aldrich syndrome;not provided
NM_032492.3(JAGN1):c.3G>A (p.Met1Ile)	JAGN1	3	9890725	Severe congenital neutropenia;Severe congenital neutropenia 6, autosomal recessive
NM_032492.3(JAGN1):c.130C>T (p.His44Tyr)	JAGN1	3	9892955	Severe congenital neutropenia;Severe congenital neutropenia 6, autosomal recessive
NM_006920.4(SCN1A):c.2923C>T (p.Leu975Phe)	SCN1A	2	166036521	Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.3970G>A (p.Val1324Met)	SCN1A	2	166002753	Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.715G>A (p.Ala239Thr)	SCN1A	2	166051968	Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.2552G>A (p.Arg851Gln)	SCN1A	2	166039427	Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.2782C>T (p.His928Tyr)	SCN1A	2	166037907	Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.4265G>A (p.Gly1422Glu)	SCN1A	2	165999763	Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.4724G>A (p.Gly1575Glu)	SCN1A	2	165994241	Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.4909C>T (p.Arg1637Cys)	SCN1A	2	165992333	Severe myoclonic epilepsy in infancy
NM_006920.4(SCN1A):c.4940C>T (p.Thr1647Met)	SCN1A	2	165992302	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.5662C>T (p.Gln1888Ter)	SCN1A	2	165991613	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.5461C>T (p.Gln1821Ter)	SCN1A	2	165991814	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4997C>T (p.Ser1666Phe)	SCN1A	2	165992278	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4985C>T (p.Ala1662Val)	SCN1A	2	165992290	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4933C>T (p.Arg1645Ter)	SCN1A	2	165992342	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4811G>A (p.Trp1604Ter)	SCN1A	2	165994187	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4412C>T (p.Ser1471Phe)	SCN1A	2	165998102	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4302G>A (p.Trp1434Ter)	SCN1A	2	165999759	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4223G>A (p.Trp1408Ter)	SCN1A	2	166002533	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4044G>A (p.Met1348Ile)	SCN1A	2	166002712	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.4016C>T (p.Ala1339Val)	SCN1A	2	166002740	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.3858G>A (p.Trp1286Ter)	SCN1A	2	166012130	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.3730C>T (p.Gln1244Ter)	SCN1A	2	166012258	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.3705+1G>A	SCN1A	2	166013743	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.3615G>A (p.Trp1205Ter)	SCN1A	2	166013834	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.3550+1G>A	SCN1A	2	166015606	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.3106C>T (p.Gln1036Ter)	SCN1A	2	166036371	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.2936G>A (p.Gly979Glu)	SCN1A	2	166037786	Severe myoclonic epilepsy in infancy
NM_001165963.1(SCN1A):c.2929G>A (p.Val977Met)	SCN1A	2	166037793	Severe myoclonic epilepsy in infancy

NM_001165963.1(SCN1A):c.2303C>T (p.Pro768Leu)	SCN1A	2	166041343	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.2213G>A (p.Trp738Ter)	SCN1A	2	166041433	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.1970C>T (p.Pro657Leu)	SCN1A	2	166043742	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.1834C>T (p.Arg612Ter)	SCN1A	2	166043878	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.1516C>T (p.Gln506Ter)	SCN1A	2	166045189	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.1259C>T (p.Ala420Val)	SCN1A	2	166046888	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.1072C>T (p.Pro358ser)	SCN1A	2	166047725	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.1025C>T (p.Ala342Val)	SCN1A	2	166048889	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.1007G>A (p.Cys336Tyr)	SCN1A	2	166048907	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.70G>A (p.Ala24Thr)	SCN1A	2	166073552	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.5436G>A (p.Trp1812Ter)	SCN1A	2	165991839	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.3657G>A (p.Trp1219Ter)	SCN1A	2	166013792	Severe myoclonic epilepsy in infancy	
NM_001165963.1(SCN1A):c.264+5G>A	SCN1A	2	166073353	Severe myoclonic epilepsy in infancy	
NM_006920.4(SCN1A):c.1178G>A (p.Arg393His)	SCN1A	2	166046969	Severe myoclonic epilepsy in infancy;not provided	
NM_006920.4(SCN1A):c.2804G>A (p.Arg935His)	SCN1A	2	166037885	Severe myoclonic epilepsy in infancy;not provided	
NM_006920.4(SCN1A):c.2816G>A (p.Gly939Glu)	SCN1A	2	166037873	Severe myoclonic epilepsy in infancy;not provided	
NM_006920.4(SCN1A):c.3701G>A (p.Arg1234Gln)	SCN1A	2	166012254	Severe myoclonic epilepsy in infancy;not provided	
NM_006920.4(SCN1A):c.4135G>A (p.Val1379Met)	SCN1A	2	166002588	Severe myoclonic epilepsy in infancy;not provided	
NM_006920.4(SCN1A):c.4901G>A (p.Arg1634Gln)	SCN1A	2	165992341	Severe myoclonic epilepsy in infancy;not provided	
NM_006920.4(SCN1A):c.5314G>A (p.Ala1772Thr)	SCN1A	2	165991928	Severe myoclonic epilepsy in infancy;not provided	
NM_006920.4(SCN1A):c.677C>T (p.Thr226Met)	SCN1A	2	166052869	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.5674C>T (p.Arg1892Ter)	SCN1A	2	165991601	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.5656C>T (p.Arg1886Ter)	SCN1A	2	165991619	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.5222G>A (p.Cys1741Tyr)	SCN1A	2	165992053	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.4573C>T (p.Arg1525Ter)	SCN1A	2	165996021	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.3818C>T (p.Ala1273Val)	SCN1A	2	166012170	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.3637C>T (p.Arg1213Ter)	SCN1A	2	166013812	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.2876G>A (p.Cys959Tyr)	SCN1A	2	166037846	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.2593C>T (p.Arg865Ter)	SCN1A	2	166038129	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.1738C>T (p.Arg580Ter)	SCN1A	2	166043974	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.1624C>T (p.Arg542Ter)	SCN1A	2	166045081	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.1129C>T (p.Arg377Ter)	SCN1A	2	166047668	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.965-1G>A	SCN1A	2	166048950	Severe myoclonic epilepsy in infancy;not provided	
NM_001165963.1(SCN1A):c.3985C>T (p.Arg1329Ter)	SCN1A	2	166009736	Severe myoclonic epilepsy in infancy;not provided	
NM_000252.2(MTM1):c.205C>T (p.Arg69Cys)	MTM1	X	150598660	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.679-1G>A	MTM1	X	150645682	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.721C>T (p.Arg241Cys)	MTM1	X	150645725	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.670C>T (p.Arg224Ter)	MTM1	X	150641410	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.469G>A (p.Glu157Lys)	MTM1	X	150638967	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.3G>A (p.Met1Ile)	MTM1	X	150592617	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.63+1G>A	MTM1	X	150592678	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.64-1G>A	MTM1	X	150596497	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.85C>T (p.Arg29Ter)	MTM1	X	150596519	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.109C>T (p.Arg37Ter)	MTM1	X	150596543	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.145G>A (p.Val49Ile)	MTM1	X	150598600	Severe X-linked myotubular myopathy	

NM_000252.2(MTM1):c.208C>T (p.Leu70Phe)	MTM1	X	150598663	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.232-1G>A	MTM1	X	150614588	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.535C>T (p.Pro179Ser)	MTM1	X	150641275	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.549G>A (p.Trp183Ter)	MTM1	X	150641289	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.557C>T (p.Thr186Ile)	MTM1	X	150641297	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.614C>T (p.Pro205Leu)	MTM1	X	150641354	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.637C>T (p.Leu213Phe)	MTM1	X	150641377	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.664C>T (p.Arg222Ter)	MTM1	X	150641404	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.678+1G>A	MTM1	X	150641419	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.679G>A (p.Val227Met)	MTM1	X	150645683	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.757C>T (p.Arg253Ter)	MTM1	X	150645761	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.867+1G>A	MTM1	X	150645872	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1132G>A (p.Gly378Arg)	MTM1	X	150657899	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1136G>A (p.Trp379Ter)	MTM1	X	150657903	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1137G>A (p.Trp379Ter)	MTM1	X	150657904	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1232G>A (p.Trp411Ter)	MTM1	X	150657999	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1244G>A (p.Gly415Glu)	MTM1	X	150658011	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1260+1G>A	MTM1	X	150658028	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1261C>T (p.Arg421Ter)	MTM1	X	150659664	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1337G>A (p.Trp446Ter)	MTM1	X	150659740	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1353+1G>A	MTM1	X	150659757	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1354-1G>A	MTM1	X	150660370	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1420C>T (p.Arg474Ter)	MTM1	X	150660437	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1456C>T (p.Arg486Ter)	MTM1	X	150660473	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1467+1G>A	MTM1	X	150660485	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.1558C>T (p.Arg520Ter)	MTM1	X	150663523	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.444+1G>A	MTM1	X	150619140	Severe X-linked myotubular myopathy	
NM_000252.2(MTM1):c.70C>T (p.Arg24Ter)	MTM1	X	150596504	Severe X-linked myotubular myopathy;not provided	
NM_000252.2(MTM1):c.231+1G>A	MTM1	X	150598687	Severe X-linked myotubular myopathy;not provided	
NM_000252.2(MTM1):c.1262G>A (p.Arg421Gln)	MTM1	X	150659665	Severe X-linked myotubular myopathy;not provided	
NM_000891.2(KCNJ2):c.514G>A (p.Asp172Asn)	KCNJ2	17	70175553	Short QT syndrome 3;short QT syndrome	
NM_001006657.1(WDR35):c.1633C>T (p.Arg545Ter)	WDR35	2	19946495	Short rib polydactyly syndrome 5	
NM_001199397.1(NEK1):c.379C>T (p.Arg127Ter)	NEK1	4	169590743	Short rib-polydactyly syndrome, Majewski type	
NM_001199397.1(NEK1):c.433G>A (p.Gly145Arg)	NEK1	4	169589478	Short rib-polydactyly syndrome, Majewski type	
NM_001199397.1(NEK1):c.117+1G>A	NEK1	4	169602513	Short rib-polydactyly syndrome, Majewski type	
NM_003995.3(NPR2):c.2455C>T (p.Arg819Cys)	NPR2	9	35806474	Short stature with nonspecific skeletal abnormalities	
NM_003995.3(NPR2):c.328C>T (p.Arg110Cys)	NPR2	9	35792736	Short stature with nonspecific skeletal abnormalities	
NM_173849.2(GSC):c.400C>T (p.Gln134Ter)	GSC	14	94769173	Short stature, auditory canal atresia, mandibular hypoplasia, and skeletal abnormalities	
NM_019023.2(PRMT7):c.1276-1G>A	PRMT7	16	68347630	SHORT STATURE, BRACHYDACTYLY, INTELLECTUAL DEVELOPMENTAL DISABILITY, AND SEIZURES	
NM_001135055.2(TKT):c.952C>T (p.Arg318Cys)	TKT	3	53230612	Short stature, developmental delay, and congenital heart defects	
NM_001135055.2(TKT):c.633G>A (p.Trp211Ter)	TKT	3	53233271	Short stature, developmental delay, and congenital heart defects	
NM_198407.2(GHSR):c.6G>A (p.Trp2Ter)	GHSR	3	172448408	Short stature, idiopathic, autosomal	
NM_000163.4(GHR):c.184G>A (p.Glu62Lys)	GHR	5	42688937	Short stature, idiopathic, autosomal	
NM_000163.4(GHR):c.484G>A (p.Val162Ile)	GHR	5	42699868	Short stature, idiopathic, autosomal	
NM_000451.3(SHOX):c.583C>T (p.Arg195Ter)	SHOX	X	641037	Short stature, idiopathic, X-linked	

NM_000451.3(SHOX):c.583C>T (p.Arg195Ter)	SHOX	Y	641037	Short stature, idiopathic, X-linked
NM_000451.3(SHOX):c.544+1G>A	SHOX	X	640879	Short stature, idiopathic, X-linked
NM_000451.3(SHOX):c.544+1G>A	SHOX	Y	640879	Short stature, idiopathic, X-linked
NM_022406.3(XRCC4):c.823C>T (p.Arg275Ter)	XRCC4	5	83258607	Short stature, microcephaly, and endocrine dysfunction
NM_022406.3(XRCC4):c.673C>T (p.Arg225Ter)	XRCC4	5	83204849	Short stature, microcephaly, and endocrine dysfunction
NM_022406.3(XRCC4):c.481C>T (p.Arg161Ter)	XRCC4	5	83195935	Short stature, microcephaly, and endocrine dysfunction
NM_022406.3(XRCC4):c.482G>A (p.Arg161Gln)	XRCC4	5	83195936	Short stature, microcephaly, and endocrine dysfunction
NM_015909.3(NBAS):c.5741G>A (p.Arg1914His)	NBAS	2	15238670	Short stature, optic nerve atrophy, and pelger-huet anomaly
NM_181523.2(PIK3R1):c.1465G>A (p.Glu489Lys)	PIK3R1	5	68294575	SHORT syndrome
NM_181523.2(PIK3R1):c.1892G>A (p.Arg631Gln)	PIK3R1	5	68296248	SHORT syndrome
NM_181523.2(PIK3R1):c.1945C>T (p.Arg649Trp)	PIK3R1	5	68296301	SHORT syndrome;not provided
NM_015662.2(IFT172):c.3907C>T (p.Arg1303Ter)	IFT172	2	27453428	Short-rib thoracic dysplasia 10 with polydactyly
NM_015662.2(IFT172):c.2716C>T (p.Gln906Ter)	IFT172	2	27459449	Short-rib thoracic dysplasia 10 without polydactyly
NM_015662.2(IFT172):c.3228+1G>A	IFT172	2	27457638	Short-rib thoracic dysplasia 10 without polydactyly
NM_015662.2(IFT172):c.886C>T (p.Arg296Trp)	IFT172	2	27480049	Short-rib thoracic dysplasia 10 without polydactyly
NM_052844.3(WDR34):c.1022C>T (p.Ala341Val)	WDR34	9	128634881	Short-rib thoracic dysplasia 11 with or without polydactyly
NM_052844.3(WDR34):c.1061C>T (p.Thr354Met)	WDR34	9	128634842	Short-rib thoracic dysplasia 11 with or without polydactyly
NM_052844.3(WDR34):c.1339C>T (p.Arg447Trp)	WDR34	9	128634259	Short-rib thoracic dysplasia 11 with or without polydactyly
NM_052844.3(WDR34):c.1340G>A (p.Arg447Gln)	WDR34	9	128634258	Short-rib thoracic dysplasia 11 with or without polydactyly
NM_052844.3(WDR34):c.1177G>A (p.Gly393Ser)	WDR34	9	128634726	Short-rib thoracic dysplasia 11 with or without polydactyly
NM_052844.3(WDR34):c.472C>T (p.Gln158Ter)	WDR34	9	128636991	Short-rib thoracic dysplasia 11 with or without polydactyly
NM_001244189.1(KIAA0586):c.1815G>A (p.Gln605=)	KIAA0586	14	58458545	Short-rib thoracic dysplasia 14 with polydactyly
NM_016008.3(DYNC2LI1):c.372G>A (p.Trp124Ter)	DYNC2LI1	2	43794508	SHORT-RIB THORACIC DYSPLASIA 15 WITH POLYDACTYLY
NM_016008.3(DYNC2LI1):c.993+1G>A	DYNC2LI1	2	43805247	SHORT-RIB THORACIC DYSPLASIA 15 WITH POLYDACTYLY
NM_001193464.1(DYNC2LI1):c.622C>T (p.Arg208Ter)	DYNC2LI1	2	43796760	SHORT-RIB THORACIC DYSPLASIA 15 WITH POLYDACTYLY
NM_001193464.1(DYNC2LI1):c.662C>T (p.Thr221Ile)	DYNC2LI1	2	43800845	SHORT-RIB THORACIC DYSPLASIA 15 WITH POLYDACTYLY
NM_016004.4(IFT52):c.424C>T (p.Arg142Ter)	IFT52	20	43605012	SHORT-RIB THORACIC DYSPLASIA 16 WITH OR WITHOUT POLYDACTYLY
NM_016004.4(IFT52):c.595G>A (p.Ala199Thr)	IFT52	20	43613959	SHORT-RIB THORACIC DYSPLASIA 16 WITH OR WITHOUT POLYDACTYLY
NM_152773.4(TCTEX1D2):c.262C>T (p.Arg88Ter)	TCTEX1D2	3	196306998	SHORT-RIB THORACIC DYSPLASIA 17 WITH OR WITHOUT POLYDACTYLY
NM_001080463.1(DYNC2H1):c.1759C>T (p.Arg587Cys)	DYNC2H1	11	103125197	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_001080463.1(DYNC2H1):c.6614G>A (p.Arg2205His)	DYNC2H1	11	103185032	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_001080463.1(DYNC2H1):c.8512C>T (p.Arg2838Ter)	DYNC2H1	11	103209933	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_001080463.1(DYNC2H1):c.7985G>A (p.Arg2662Gln)	DYNC2H1	11	103199373	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_001080463.1(DYNC2H1):c.7486C>T (p.Pro2496Ser)	DYNC2H1	11	103191565	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_001080463.1(DYNC2H1):c.988C>T (p.Arg330Cys)	DYNC2H1	11	103117852	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_001080463.1(DYNC2H1):c.12177+1G>A	DYNC2H1	11	103358360	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_018051.4(WDR60):c.1891C>T (p.Gln631Ter)	WDR60	7	158918839	Short-rib thoracic dysplasia 8 with or without polydactyly
NM_018051.4(WDR60):c.2246C>T (p.Thr749Met)	WDR60	7	158923722	Short-rib thoracic dysplasia 8 with or without polydactyly
NM_014714.3(IFT140):c.1565G>A (p.Gly522Glu)	IFT140	16	1571494	Short-rib thoracic dysplasia without polydactyly
NM_015634.3(KIF1BP):c.268C>T (p.Arg90Ter)	KIF1BP	10	68989100	Sprintzen-Goldberg syndrome
NM_003036.3(SKI):c.347G>A (p.Gly116Glu)	SKI	1	2229113	Sprintzen-Goldberg syndrome
NM_003036.3(SKI):c.101G>A (p.Gly34Asp)	SKI	1	2228867	Sprintzen-Goldberg syndrome
NM_003036.3(SKI):c.103C>T (p.Pro35Ser)	SKI	1	2228869	Sprintzen-Goldberg syndrome
NM_003036.3(SKI):c.100G>A (p.Gly34Ser)	SKI	1	2228866	Sprintzen-Goldberg syndrome;not provided
NM_016038.3(SBDS):c.505C>T (p.Arg169Cys)	SBDS	7	66991256	Shwachman syndrome

NM_016038.3(SBDS):c.652C>T (p.Arg218Ter)	SBDS	7	66988472	Shwachman syndrome
NM_015697.7(COQ2):c.1160G>A (p.Arg387Gln)	COQ2	4	83264305	Shy-Drager syndrome
NM_000434.3(NEU1):c.946C>T (p.Pro316Ser)	NEU1	6	31860117	Sialidosis type I
NM_000434.3(NEU1):c.727G>A (p.Gly243Arg)	NEU1	6	31860510	Sialidosis type I;not provided
NM_000434.3(NEU1):c.649G>A (p.Val217Met)	NEU1	6	31860588	Sialidosis type I;Sialidosis, type II;not provided
NM_000434.3(NEU1):c.87G>A (p.Trp29Ter)	NEU1	6	31862690	Sialidosis, type II
NM_000434.3(NEU1):c.239C>T (p.Pro80Leu)	NEU1	6	31862112	Sialidosis, type II
NM_000434.3(NEU1):c.893C>T (p.Ala298Val)	NEU1	6	31860170	Sialidosis, type II
NM_000434.3(NEU1):c.69G>A (p.Trp23Ter)	NEU1	6	31862708	Sialidosis, type II
NM_005476.5(GNE):c.796C>T (p.Arg266Trp)	GNE	9	36234106	Sialuria
NM_005476.5(GNE):c.797G>A (p.Arg266Gln)	GNE	9	36234105	Sialuria
NM_005477.2(HCN4):c.1657G>A (p.Asp553Asn)	HCN4	15	73325378	Sick sinus syndrome 2, autosomal dominant
NM_002471.3(MYH6):c.2161C>T (p.Arg721Trp)	MYH6	14	23396970	Sick sinus syndrome 3, susceptibility to
NM_000335.4(SCN5A):c.3890C>T (p.Pro1297Leu)	SCN5A	3	38562485	Sick sinus syndrome;Sick sinus syndrome 1, autosomal recessive
NM_015107.2(PHF8):c.631C>T (p.Arg211Ter)	PHF8	X	54014529	Siderius X-linked mental retardation syndrome
NM_001302946.1(TRNT1):c.461C>T (p.Thr154Ile)	TRNT1	3	3140628	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay
NM_001273.3(CHD4):c.4822G>A (p.Val1608Ile)	CHD4	12	6581131	SIFRIM-HITZ-WEISS SYNDROME
NM_001273.3(CHD4):c.3203G>A (p.Arg1068His)	CHD4	12	6591713	SIFRIM-HITZ-WEISS SYNDROME
NM_001273.3(CHD4):c.3380G>A (p.Arg1127Gln)	CHD4	12	6588383	SIFRIM-HITZ-WEISS SYNDROME
NM_032667.6(BSCL2):c.269C>T (p.Ser90Leu)	BSCL2	11	62702493	Silver spastic paraplegia syndrome;not provided
NM_004484.3(GPC3):c.595C>T (p.Arg199Ter)	GPC3	X	133753919	Simpson-Golabi-Behmel syndrome
NM_004484.3(GPC3):c.337+1G>A	GPC3	X	133953049	Simpson-Golabi-Behmel syndrome
NM_004484.3(GPC3):c.1159C>T (p.Arg387Ter)	GPC3	X	133699902	Simpson-Golabi-Behmel syndrome
NM_004484.3(GPC3):c.1666G>A (p.Gly556Arg)	GPC3	X	133536201	Simpson-Golabi-Behmel syndrome
NM_022168.3(IFIH1):c.2465G>A (p.Arg822Gln)	IFIH1	2	162272377	Singleton-Merten syndrome 1;not provided
NM_000720.3(CACNA1D):c.1127C>T (p.Ala376Val)	CACNA1D	3	53673723	Sinoatrial node dysfunction and deafness
NM_022437.2(ABCG8):c.1720G>A (p.Gly574Arg)	ABCG8	2	43875377	Sitosterolemia
NM_022437.2(ABCG8):c.788G>A (p.Arg263Gln)	ABCG8	2	43852692	Sitosterolemia
NM_022437.2(ABCG8):c.1234C>T (p.Arg412Ter)	ABCG8	2	43873809	Sitosterolemia
NM_022436.2(ABCG5):c.1222C>T (p.Arg408Ter)	ABCG5	2	43824015	Sitosterolemia
NM_022436.2(ABCG5):c.727C>T (p.Arg243Ter)	ABCG5	2	43826429	Sitosterolemia
NM_022436.2(ABCG5):c.1256G>A (p.Arg419His)	ABCG5	2	43823981	Sitosterolemia
NM_022436.2(ABCG5):c.1166G>A (p.Arg389His)	ABCG5	2	43824071	Sitosterolemia
NM_022436.2(ABCG5):c.46C>T (p.Gln16Ter)	ABCG5	2	43838634	Sitosterolemia
NM_022436.2(ABCG5):c.1336C>T (p.Arg446Ter)	ABCG5	2	43822924	Sitosterolemia
NM_022437.2(ABCG8):c.1083G>A (p.Trp361Ter)	ABCG8	2	43872094	Sitosterolemia;not provided
NM_000382.2(ALDH3A2):c.641G>A (p.Cys214Tyr)	ALDH3A2	17	19656535	SjÃ¶gren-Larsson syndrome
NM_000382.2(ALDH3A2):c.943C>T (p.Pro315Ser)	ALDH3A2	17	19663335	SjÃ¶gren-Larsson syndrome;not provided
NM_000382.2(ALDH3A2):c.551C>T (p.Thr184Met)	ALDH3A2	17	19656445	SjÃ¶gren-Larsson syndrome;not provided
NM_021625.4(TRPV4):c.1875G>A (p.Met625Ile)	TRPV4	12	109792379	Skeletal dysplasia
NM_021625.4(TRPV4):c.2330G>A (p.Cys777Tyr)	TRPV4	12	109786716	Skeletal dysplasia
NM_021625.4(TRPV4):c.547G>A (p.Glu183Lys)	TRPV4	12	109808308	Skeletal dysplasia;Spondyloepiphyseal dysplasia Maroteaux type
NM_001143826.2(MAPRE2):c.325C>T (p.Gln109Ter)	MAPRE2	18	35102003	Skin creases, congenital symmetric circumferential, 2
NM_001143826.2(MAPRE2):c.298C>T (p.Arg100Cys)	MAPRE2	18	35101976	Skin creases, congenital symmetric circumferential, 2
NM_004415.3(DSP):c.7096C>T (p.Arg2366Cys)	DSP	6	7584358	Skin fragility woolly hair syndrome

NM_004415.3(DSP):c.1990C>T (p.Gln664Ter)	DSP	6	7571928	Skin fragility woolly hair syndrome
NM_004415.3(DSP):c.7097G>A (p.Arg2366His)	DSP	6	7584359	Skin fragility woolly hair syndrome
NM_014629.3(ARHGEF10):c.995C>T (p.Thr332Ile)	ARHGEF10	8	1882669	Slowed nerve conduction velocity, autosomal dominant
NM_002977.3(SCN9A):c.1867G>A (p.Asp623Asn)	SCN9A	2	166284560	Small fiber neuropathy
NM_004958.3(MTOR):c.4785G>A (p.Met1595Ile)	MTOR	1	11144735	Smith-Kingsmore syndrome
NM_004958.3(MTOR):c.6981G>A (p.Met2327Ile)	MTOR	1	11117039	Smith-Kingsmore syndrome
NM_004958.3(MTOR):c.5395G>A (p.Glu1799Lys)	MTOR	1	11130747	Smith-Kingsmore syndrome;not provided
NM_001360.2(DHCR7):c.730G>A (p.Gly244Arg)	DHCR7	11	71438980	Smith-Lemli-Opitz syndrome
NM_001360.2(DHCR7):c.453G>A (p.Trp151Ter)	DHCR7	11	71441400	Smith-Lemli-Opitz syndrome
NM_001360.2(DHCR7):c.1210C>T (p.Arg404Cys)	DHCR7	11	71435593	Smith-Lemli-Opitz syndrome
NM_001360.2(DHCR7):c.3G>A (p.Met1Ile)	DHCR7	11	71444950	Smith-Lemli-Opitz syndrome
NM_001360.2(DHCR7):c.506C>T (p.Ser169Leu)	DHCR7	11	71441347	Smith-Lemli-Opitz syndrome
NM_001360.2(DHCR7):c.725G>A (p.Arg242His)	DHCR7	11	71438985	Smith-Lemli-Opitz syndrome
NM_001360.2(DHCR7):c.907G>A (p.Gly303Arg)	DHCR7	11	71437868	Smith-Lemli-Opitz syndrome
NM_001163817.1(DHCR7):c.1328G>A (p.Arg443His)	DHCR7	11	71435475	Smith-Lemli-Opitz syndrome
NM_001360.2(DHCR7):c.1054C>T (p.Arg352Trp)	DHCR7	11	71435749	Smith-Lemli-Opitz syndrome;not provided
NM_001360.2(DHCR7):c.866C>T (p.Thr289Ile)	DHCR7	11	71437909	Smith-Lemli-Opitz syndrome;not provided
NM_001360.2(DHCR7):c.1342G>A (p.Glu448Lys)	DHCR7	11	71435461	Smith-Lemli-Opitz syndrome;not provided
NM_001360.2(DHCR7):c.1055G>A (p.Arg352Gln)	DHCR7	11	71435748	Smith-Lemli-Opitz syndrome;not provided
NM_001360.2(DHCR7):c.1228G>A (p.Gly410Ser)	DHCR7	11	71435575	Smith-Lemli-Opitz syndrome;not provided
NM_001360.2(DHCR7):c.724C>T (p.Arg242Cys)	DHCR7	11	71438986	Smith-Lemli-Opitz syndrome;not provided
NM_001360.2(DHCR7):c.151C>T (p.Pro51Ser)	DHCR7	11	71444163	Smith-Lemli-Opitz syndrome;not provided
NM_001360.2(DHCR7):c.841G>A (p.Val281Met)	DHCR7	11	71437934	Smith-Lemli-Opitz syndrome;not provided
NM_030665.3(RAI1):c.5423G>A (p.Ser1808Asn)	RAI1	17	17798371	Smith-Magenis syndrome
NM_030665.3(RAI1):c.2273G>A (p.Trp758Ter)	RAI1	17	17795221	Smith-Magenis syndrome
NM_030665.3(RAI1):c.4678C>T (p.Arg1560Ter)	RAI1	17	17797626	Smith-Magenis syndrome;not provided
NM_017653.3(DYM):c.259G>A (p.Glu87Lys)	DYM	18	49379693	Smith-McCort dysplasia 1
NM_002242.4(KCNJ13):c.484C>T (p.Arg162Trp)	KCNJ13	2	232768790	Snowflake vitreoretinal degeneration
NM_004595.4(SMS):c.329+5G>A	SMS	X	21972576	Snyder Robinson syndrome
NM_004595.4(SMS):c.200G>A (p.Gly67Glu)	SMS	X	21971926	Snyder Robinson syndrome
NM_001172560.1(SSTR5):c.718C>T (p.Arg240Trp)	SSTR5	16	1079586	Somatostatin analog, resistance to
NM_003977.3(AIP):c.469-1G>A	AIP	11	67490037	Somatotroph adenoma
NM_003977.3(AIP):c.910C>T (p.Arg304Ter)	AIP	11	67490910	Somatotroph adenoma
NM_003977.3(AIP):c.64C>T (p.Arg22Ter)	AIP	11	67483222	Somatotroph adenoma
NM_003977.3(AIP):c.241C>T (p.Arg81Ter)	AIP	11	67487147	Somatotroph adenoma
NM_003977.3(AIP):c.811C>T (p.Arg271Trp)	AIP	11	67490811	Somatotroph adenoma
NM_022455.4(NSD1):c.6605G>A (p.Cys2202Tyr)	NSD1	5	177293973	Sotos syndrome 1
NM_022455.4(NSD1):c.1262G>A (p.Trp421Ter)	NSD1	5	177209661	Sotos syndrome 1
NM_022455.4(NSD1):c.3067C>T (p.Arg1023Ter)	NSD1	5	177211466	Sotos syndrome 1
NM_022455.4(NSD1):c.3214C>T (p.Arg1072Ter)	NSD1	5	177211613	Sotos syndrome 1
NM_022455.4(NSD1):c.3839G>A (p.Trp1280Ter)	NSD1	5	177235863	Sotos syndrome 1
NM_022455.4(NSD1):c.3964C>T (p.Arg1322Ter)	NSD1	5	177238279	Sotos syndrome 1
NM_022455.4(NSD1):c.4378+1G>A	NSD1	5	177244271	Sotos syndrome 1
NM_022455.4(NSD1):c.4417C>T (p.Arg1473Ter)	NSD1	5	177246716	Sotos syndrome 1
NM_022455.4(NSD1):c.4444C>T (p.Gln1482Ter)	NSD1	5	177246743	Sotos syndrome 1

NM_022455.4(NSD1):c.4681C>T (p.Gln1561Ter)	NSD1	5	177251769	Sotos syndrome 1
NM_022455.4(NSD1):c.4907C>T (p.Ser1636Phe)	NSD1	5	177257092	Sotos syndrome 1
NM_022455.4(NSD1):c.4966+1G>A	NSD1	5	177257152	Sotos syndrome 1
NM_022455.4(NSD1):c.4987C>T (p.Arg1663Cys)	NSD1	5	177260009	Sotos syndrome 1
NM_022455.4(NSD1):c.5098C>T (p.Arg1700Ter)	NSD1	5	177260120	Sotos syndrome 1
NM_022455.4(NSD1):c.5146+1G>A	NSD1	5	177260169	Sotos syndrome 1
NM_022455.4(NSD1):c.5309G>A (p.Trp1770Ter)	NSD1	5	177269607	Sotos syndrome 1
NM_022455.4(NSD1):c.5375G>A (p.Gly1792Glu)	NSD1	5	177269673	Sotos syndrome 1
NM_022455.4(NSD1):c.5425C>T (p.Gln1809Ter)	NSD1	5	177269723	Sotos syndrome 1
NM_022455.4(NSD1):c.5431C>T (p.Arg1811Ter)	NSD1	5	177269729	Sotos syndrome 1
NM_022455.4(NSD1):c.5432G>A (p.Arg1811Gln)	NSD1	5	177269730	Sotos syndrome 1
NM_022455.4(NSD1):c.5566C>T (p.Gln1856Ter)	NSD1	5	177273728	Sotos syndrome 1
NM_022455.4(NSD1):c.5740C>T (p.Arg1914Cys)	NSD1	5	177280682	Sotos syndrome 1
NM_022455.4(NSD1):c.5800C>T (p.Gln1934Ter)	NSD1	5	177280742	Sotos syndrome 1
NM_022455.4(NSD1):c.5965C>T (p.Gln1989Ter)	NSD1	5	177282537	Sotos syndrome 1
NM_022455.4(NSD1):c.6049C>T (p.Arg2017Trp)	NSD1	5	177283826	Sotos syndrome 1
NM_022455.4(NSD1):c.6070C>T (p.Gln2024Ter)	NSD1	5	177283847	Sotos syndrome 1
NM_022455.4(NSD1):c.6258+1G>A	NSD1	5	177288926	Sotos syndrome 1
NM_022455.4(NSD1):c.6307C>T (p.Gln2103Ter)	NSD1	5	177292002	Sotos syndrome 1
NM_022455.4(NSD1):c.6310C>T (p.Gln2104Ter)	NSD1	5	177292005	Sotos syndrome 1
NM_022455.4(NSD1):c.6455G>A (p.Arg2152Gln)	NSD1	5	177292150	Sotos syndrome 1
NM_022455.4(NSD1):c.6476G>A (p.Cys2159Tyr)	NSD1	5	177293844	Sotos syndrome 1
NM_022455.4(NSD1):c.6559C>T (p.Arg2187Ter)	NSD1	5	177293927	Sotos syndrome 1
NM_022455.4(NSD1):c.6656G>A (p.Arg2219His)	NSD1	5	177294024	Sotos syndrome 1
NM_022455.4(NSD1):c.5332C>T (p.Arg1778Ter)	NSD1	5	177269630	Sotos syndrome 1
NM_022455.4(NSD1):c.5950C>T (p.Arg1984Ter)	NSD1	5	177282522	Sotos syndrome 1
NM_022455.4(NSD1):c.7966C>T (p.Gln2656Ter)	NSD1	5	177295334	Sotos syndrome 1
NM_022455.4(NSD1):c.6152-1G>A	NSD1	5	177288818	Sotos syndrome 1
NM_022455.4(NSD1):c.3958C>T (p.Arg1320Ter)	NSD1	5	177238273	Sotos syndrome 1;not provided
NM_022455.4(NSD1):c.6463+1G>A	NSD1	5	177292159	Sotos syndrome 1;not provided
NM_022455.4(NSD1):c.1831C>T (p.Arg611Ter)	NSD1	5	177210230	Sotos syndrome 1;not provided
NM_022455.4(NSD1):c.3091C>T (p.Arg1031Ter)	NSD1	5	177211490	Sotos syndrome 1;not provided
NM_022455.4(NSD1):c.5951G>A (p.Arg1984Gln)	NSD1	5	177282523	Sotos syndrome 1;not provided
NM_022455.4(NSD1):c.6013C>T (p.Arg2005Ter)	NSD1	5	177283790	Sotos syndrome 1;not provided
NM_022455.4(NSD1):c.6014G>A (p.Arg2005Gln)	NSD1	5	177283791	Sotos syndrome 1;not provided
NM_022455.4(NSD1):c.6454C>T (p.Arg2152Ter)	NSD1	5	177292149	Sotos syndrome 1;not provided
NM_002501.3(NFIX):c.568C>T (p.Gln190Ter)	NFIX	19	13073055	Sotos syndrome 2
NM_006796.2(AFG3L2):c.1875G>A (p.Met625Ile)	AFG3L2	18	12340306	Spastic ataxia 5, autosomal recessive
NM_014363.5(SACS):c.7504C>T (p.Arg2502Ter)	SACS	13	23336372	Spastic ataxia Charlevoix-Saguenay type
NM_014363.5(SACS):c.10907G>A (p.Arg3636Gln)	SACS	13	23332969	Spastic ataxia Charlevoix-Saguenay type
NM_014363.5(SACS):c.12160C>T (p.Gln4054Ter)	SACS	13	23331716	Spastic ataxia Charlevoix-Saguenay type
NM_014363.5(SACS):c.10906C>T (p.Arg3636Ter)	SACS	13	23332970	Spastic ataxia Charlevoix-Saguenay type
NM_014363.5(SACS):c.12028C>T (p.Gln4010Ter)	SACS	13	23331848	Spastic ataxia Charlevoix-Saguenay type
NM_014363.5(SACS):c.5125C>T (p.Gln1709Ter)	SACS	13	23338751	Spastic ataxia Charlevoix-Saguenay type
NM_014363.5(SACS):c.9508C>T (p.Arg3170Ter)	SACS	13	23334368	Spastic ataxia Charlevoix-Saguenay type;not provided

NM_014946.3(SPAST):c.1495C>T (p.Arg499Cys)	SPAST	2	32141905	Spastic paraparesis;Spastic paraplegia 4, autosomal dominant
NM_004820.4(CYP7B1):c.525G>A (p.Trp175Ter)	CYP7B1	8	64616016	Spastic paraplegia
NM_014363.5(SACS):c.11707C>T (p.Arg3903Ter)	SACS	13	23332169	Spastic paraplegia
NM_000425.4(L1CAM):c.1792G>A (p.Asp598Asn)	L1CAM	X	153866034	Spastic paraplegia 1
NM_000425.4(L1CAM):c.1108G>A (p.Gly370Arg)	L1CAM	X	153869818	Spastic paraplegia 1;not provided
NM_000425.4(L1CAM):c.3581C>T (p.Ser1194Leu)	L1CAM	X	153862856	Spastic paraplegia 1,X-linked hydrocephalus syndrome;not provided
NM_004984.2(KIF5A):c.838C>T (p.Arg280Cys)	KIF5A	12	57569274	Spastic paraplegia 10
NM_004984.2(KIF5A):c.1082C>T (p.Ala361Val)	KIF5A	12	57569648	Spastic paraplegia 10
NM_004984.2(KIF5A):c.2263G>A (p.Glu755Lys)	KIF5A	12	57576825	Spastic paraplegia 10
NM_004984.2(KIF5A):c.839G>A (p.Arg280His)	KIF5A	12	57569275	Spastic paraplegia 10
NM_004984.2(KIF5A):c.704G>A (p.Gly235Glu)	KIF5A	12	57567608	Spastic paraplegia 10
NM_004984.2(KIF5A):c.694G>A (p.Asp232Asn)	KIF5A	12	57567598	Spastic paraplegia 10
NM_025137.3(SPG11):c.1951C>T (p.Arg651Ter)	SPG11	15	44628785	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.2316+1G>A	SPG11	15	44622727	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.2697G>A (p.Trp899Ter)	SPG11	15	44620327	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.4846C>T (p.Gln1616Ter)	SPG11	15	44589312	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.5470C>T (p.Arg1824Ter)	SPG11	15	44584210	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.5977C>T (p.Gln1993Ter)	SPG11	15	44574931	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.6091C>T (p.Arg2031Ter)	SPG11	15	44573661	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.6157G>A (p.Val2053Met)	SPG11	15	44573595	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.6856C>T (p.Arg2286Ter)	SPG11	15	44565997	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.869+1G>A	SPG11	15	44657094	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.1492C>T (p.Gln498Ter)	SPG11	15	44648976	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.5158C>T (p.Gln1720Ter)	SPG11	15	44584522	Spastic paraplegia 11, autosomal recessive
NM_025137.3(SPG11):c.5623C>T (p.Gln1875Ter)	SPG11	15	44584057	Spastic paraplegia 11, autosomal recessive;not provided
NM_005619.4(RTN2):c.1100C>T (p.Ser367Phe)	RTN2	19	45489487	Spastic paraplegia 12
NM_199440.1(HSPD1):c.292G>A (p.Val98Ile)	HSPD1	2	197497275	Spastic paraplegia 13
NM_015346.3(ZFYVE26):c.4312C>T (p.Arg1438Ter)	ZFYVE26	14	67782840	Spastic paraplegia 15
NM_015346.3(ZFYVE26):c.1477C>T (p.Gln493Ter)	ZFYVE26	14	67802241	Spastic paraplegia 15
NM_015346.3(ZFYVE26):c.5422C>T (p.Gln1808Ter)	ZFYVE26	14	67772109	Spastic paraplegia 15
NM_015346.3(ZFYVE26):c.4181G>A (p.Trp1394Ter)	ZFYVE26	14	67782971	Spastic paraplegia 15
NM_001128834.2(PLP1):c.418C>T (p.His140Tyr)	PLP1	X	103786691	Spastic paraplegia 2
NM_001128834.2(PLP1):c.509C>T (p.Ser170Phe)	PLP1	X	103787853	Spastic paraplegia 2
NM_001128834.2(PLP1):c.409C>T (p.Arg137Trp)	PLP1	X	103786682	Spastic paraplegia 2
NM_001478.4(B4GALNT1):c.682C>T (p.Arg228Ter)	B4GALNT1	12	57630182	Spastic paraplegia 26
NM_001478.4(B4GALNT1):c.358C>T (p.Gln120Ter)	B4GALNT1	12	57631225	Spastic paraplegia 26
NM_001127713.1(ATL1):c.650G>A (p.Arg217Gln)	ATL1	14	50613278	Spastic paraplegia 3
NM_015915.4(ATL1):c.467C>T (p.Thr156Ile)	ATL1	14	50591584	Spastic paraplegia 3
NM_001127713.1(ATL1):c.1246C>T (p.Arg416Cys)	ATL1	14	50628157	Spastic paraplegia 3
NM_015915.4(ATL1):c.715C>T (p.Arg239Cys)	ATL1	14	50613343	Spastic paraplegia 3;not provided
NM_015915.4(ATL1):c.1244G>A (p.Arg415Gln)	ATL1	14	50628155	Spastic paraplegia 3;not provided
NM_015915.4(ATL1):c.757G>A (p.Val253Ile)	ATL1	14	50614406	Spastic paraplegia 3;not provided
NM_015915.4(ATL1):c.1483C>T (p.Arg495Trp)	ATL1	14	50628394	Spastic paraplegia 3;not provided
NM_001244008.1(KIF1A):c.764C>T (p.Ala255Val)	KIF1A	2	240783773	Spastic paraplegia 30, autosomal recessive
NM_001164730.1(REEP1):c.358C>T (p.Arg120Ter)	REEP1	2	86252037	Spastic paraplegia 31, autosomal dominant

NM_022912.2(REEP1):c.595+1G>A	REEP1	2	86232624	Spastic paraplegia 31, autosomal dominant
NM_022912.2(REEP1):c.113G>A (p.Trp38Ter)	REEP1	2	86264034	Spastic paraplegia 31, autosomal dominant
NM_024306.4(FA2H):c.703C>T (p.Arg235Cys)	FA2H	16	74719071	Spastic paraplegia 35
NM_024306.4(FA2H):c.460C>T (p.Arg154Cys)	FA2H	16	74727290	Spastic paraplegia 35;not provided
NM_006702.4(PNPLA6):c.2669G>A (p.Arg890His)	PNPLA6	19	7555041	Spastic paraplegia 39
NM_001166111.1(PNPLA6):c.787G>A (p.Val263Ile)	PNPLA6	19	7540675	Spastic paraplegia 39
NM_006702.4(PNPLA6):c.2375G>A (p.Gly792Glu)	PNPLA6	19	7554578	Spastic paraplegia 39
NM_014946.3(SPAST):c.1343G>A (p.Cys448Tyr)	SPAST	2	32136898	Spastic paraplegia 4, autosomal dominant
NM_014946.3(SPAST):c.1291C>T (p.Arg431Ter)	SPAST	2	32136608	Spastic paraplegia 4, autosomal dominant
NM_014946.3(SPAST):c.1417C>T (p.Gln473Ter)	SPAST	2	32137112	Spastic paraplegia 4, autosomal dominant
NM_014946.3(SPAST):c.1685G>A (p.Arg562Gln)	SPAST	2	32145005	Spastic paraplegia 4, autosomal dominant
NM_199436.1(SPAST):c.1149+1G>A	SPAST	2	32128480	Spastic paraplegia 4, autosomal dominant
NM_014946.3(SPAST):c.1378C>T (p.Arg460Cys)	SPAST	2	32136933	Spastic paraplegia 4, autosomal dominant
NM_014946.3(SPAST):c.1173+1G>A	SPAST	2	32127023	Spastic paraplegia 4, autosomal dominant
NM_012229.4(NT5C2):c.85C>T (p.Arg29Ter)	NT5C2	10	103174874	Spastic paraplegia 45, autosomal recessive
NM_012229.4(NT5C2):c.175+1G>A	NT5C2	10	103139405	Spastic paraplegia 45, autosomal recessive
NM_020944.2(GBA2):c.1888C>T (p.Arg630Trp)	GBA2	9	35738811	Spastic paraplegia 46, autosomal recessive
NM_020944.2(GBA2):c.700C>T (p.Arg234Ter)	GBA2	9	35741758	Spastic paraplegia 46, autosomal recessive
NM_020944.2(GBA2):c.1018C>T (p.Arg340Ter)	GBA2	9	35740833	Spastic paraplegia 46, autosomal recessive
NM_020944.2(GBA2):c.2618G>A (p.Arg873His)	GBA2	9	35737335	Spastic paraplegia 46, autosomal recessive
NM_014855.2(AP5Z1):c.1322G>A (p.Trp441Ter)	AP5Z1	7	4787644	Spastic paraplegia 48, autosomal recessive
NM_014855.2(AP5Z1):c.412C>T (p.Arg138Ter)	AP5Z1	7	4783361	Spastic paraplegia 48, autosomal recessive
NM_014855.2(AP5Z1):c.616C>T (p.Arg206Trp)	AP5Z1	7	4783793	Spastic paraplegia 48, autosomal recessive
NM_014855.2(AP5Z1):c.1732C>T (p.Gln578Ter)	AP5Z1	7	4789856	Spastic paraplegia 48, autosomal recessive
NM_004722.3(AP4M1):c.577G>A (p.Glu193Lys)	AP4M1	7	100104125	Spastic paraplegia 50, autosomal recessive
NM_004722.3(AP4M1):c.1012C>T (p.Arg338Ter)	AP4M1	7	100106278	Spastic paraplegia 50, autosomal recessive
NM_001254729.1(AP4S1):c.124C>T (p.Arg42Ter)	AP4S1	14	31066320	Spastic paraplegia 52, autosomal recessive
NM_007077.4(AP4S1):c.289C>T (p.Arg97Ter)	AP4S1	14	31072968	Spastic paraplegia 52, autosomal recessive;not provided
NM_015214.2(DDHD2):c.1546C>T (p.Arg516Ter)	DDHD2	8	38252216	Spastic paraplegia 54, autosomal recessive
NM_001164232.1(DDHD2):c.859C>T (p.Arg287Ter)	DDHD2	8	38245752	Spastic paraplegia 54, autosomal recessive
NM_152269.4(C12orf65):c.394C>T (p.Arg132Ter)	C12orf65	12	123256924	Spastic paraplegia 55, autosomal recessive
NM_152269.4(C12orf65):c.415C>T (p.Gln139Ter)	C12orf65	12	123256945	Spastic paraplegia 55, autosomal recessive
NM_183075.2(CYP2U1):c.1462C>T (p.Arg488Trp)	CYP2U1	4	107950250	Spastic paraplegia 56, autosomal recessive
NM_001007565.2(TFG):c.316C>T (p.Arg106Cys)	TFG	3	100728759	Spastic paraplegia 57, autosomal recessive
NM_004820.4(CYP7B1):c.1088C>T (p.Ser363Phe)	CYP7B1	8	64604827	Spastic paraplegia 5A
NM_004820.4(CYP7B1):c.169G>A (p.Gly57Arg)	CYP7B1	8	64624493	Spastic paraplegia 5A
NM_144599.4(NIPA1):c.316G>A (p.Gly106Arg)	NIPA1	15	22812252	Spastic paraplegia 6
NM_006459.3(ERLIN1):c.763C>T (p.Arg255Ter)	ERLIN1	10	100154922	Spastic paraplegia 62, autosomal recessive
NM_003119.3(SPG7):c.1408C>T (p.Arg470Ter)	SPG7	16	89544731	Spastic paraplegia 7
NM_003119.3(SPG7):c.988-1G>A	SPG7	16	89531903	Spastic paraplegia 7
NM_003119.3(SPG7):c.1996G>A (p.Gly666Arg)	SPG7	16	89553853	Spastic paraplegia 7
NM_003119.3(SPG7):c.2249C>T (p.Pro750Leu)	SPG7	16	89556954	Spastic paraplegia 7
NM_003119.3(SPG7):c.637C>T (p.Arg213Ter)	SPG7	16	89526347	Spastic paraplegia 7
NM_003119.3(SPG7):c.1715C>T (p.Ala572Val)	SPG7	16	89550545	Spastic paraplegia 7;not provided
NM_001199752.2(CPT1C):c.109C>T (p.Arg37Cys)	CPT1C	19	49692361	Spastic paraplegia 73, autosomal dominant

NM_001198868.1(CAPN1):c.1579C>T (p.Gln527Ter)	CAPN1	11	65206793	Spastic paraplegia 76, autosomal recessive
NM_001198868.1(CAPN1):c.1605+5G>A	CAPN1	11	65206824	Spastic paraplegia 76, autosomal recessive
NM_022089.3(ATP13A2):c.3418C>T (p.Gln1140Ter)	ATP13A2	1	16986346	SPASTIC PARAPLEGIA 78, AUTOSOMAL RECESSIVE
NM_022089.3(ATP13A2):c.1550C>T (p.Thr517Ile)	ATP13A2	1	16993828	SPASTIC PARAPLEGIA 78, AUTOSOMAL RECESSIVE
NM_022089.3(ATP13A2):c.364C>T (p.Gln122Ter)	ATP13A2	1	17004805	SPASTIC PARAPLEGIA 78, AUTOSOMAL RECESSIVE
NM_022089.3(ATP13A2):c.1345C>T (p.Arg449Ter)	ATP13A2	1	16996262	SPASTIC PARAPLEGIA 78, AUTOSOMAL RECESSIVE
NM_002860.3(ALDH18A1):c.755G>A (p.Arg252Gln)	ALDH18A1	10	95633012	Spastic paraplegia 9
NM_020771.3(HACE1):c.655C>T (p.Arg219Ter)	HACE1	6	104796988	Spastic paraplegia and psychomotor retardation with or without seizures
NM_020771.3(HACE1):c.2242C>T (p.Arg748Ter)	HACE1	6	104750442	Spastic paraplegia and psychomotor retardation with or without seizures
NM_020771.3(HACE1):c.454C>T (p.Gln152Ter)	HACE1	6	104833122	Spastic paraplegia and psychomotor retardation with or without seizures
NM_020771.3(HACE1):c.805C>T (p.Arg269Ter)	HACE1	6	104796666	Spastic paraplegia and psychomotor retardation with or without seizures
NM_020738.3(KIDINS220):c.4050G>A (p.Trp1350Ter)	KIDINS220	2	8733447	SPASTIC PARAPLEGIA, INTELLECTUAL DISABILITY, NYSTAGMUS, AND OBESITY
NM_020738.3(KIDINS220):c.4096C>T (p.Gln1366Ter)	KIDINS220	2	8731940	SPASTIC PARAPLEGIA, INTELLECTUAL DISABILITY, NYSTAGMUS, AND OBESITY
NM_025137.3(SPG11):c.2146C>T (p.Gln716Ter)	SPG11	15	44626429	Spastic Paraplegia, Recessive;Spastic paraplegia 11, autosomal recessive;not provided
NM_003119.3(SPG7):c.1045G>A (p.Gly349Ser)	SPG7	16	89531961	Spastic Paraplegia, Recessive;Spastic paraplegia 7;not provided
NM_003119.3(SPG7):c.1529C>T (p.Ala510Val)	SPG7	16	89546737	Spastic Paraplegia, Recessive;Spastic paraplegia 7;not provided
NM_004321.7(KIF1A):c.38G>A (p.Arg13His)	KIF1A	2	240797715	Spastic paraplegia;not provided
NM_000425.4(L1CAM):c.807-5G>A	L1CAM	X	153870246	Spastic paraplegia;not provided
NM_004984.2(KIF5A):c.751G>A (p.Glu251Lys)	KIF5A	12	57568999	Spastic paraplegia;Spastic paraplegia 10
NM_004984.2(KIF5A):c.611G>A (p.Arg204Gln)	KIF5A	12	57567515	Spastic paraplegia;Spastic paraplegia 10
NM_004820.4(CYP7B1):c.1250G>A (p.Arg417His)	CYP7B1	8	64596913	Spastic paraplegia;Spastic paraplegia 5A;not provided
NM_003038.4(SLC1A4):c.766G>A (p.Glu256Lys)	SLC1A4	2	65010729	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly
NM_003038.4(SLC1A4):c.1369C>T (p.Arg457Trp)	SLC1A4	2	65020916	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly;not provided
NM_014491.3(FOXP2):c.1658G>A (p.Arg553His)	FOXP2	7	114662075	Speech-language disorder 1
NM_014491.3(FOXP2):c.982C>T (p.Arg328Ter)	FOXP2	7	114642616	Speech-language disorder 1
NM_144605.4(SEPT12):c.266C>T (p.Thr89Met)	12-Sep	16	4786006	Spermatogenic failure 10
NM_144605.4(SEPT12):c.589G>A (p.Asp197Asn)	12-Sep	16	4783690	Spermatogenic failure 10
NM_001329595.1(KLHL10):c.937G>A (p.Ala313Thr)	KLHL10	17	41845378	Spermatogenic failure 11
NM_005640.2(TAF4B):c.1831C>T (p.Arg611Ter)	TAF4B	18	26293530	Spermatogenic failure 13
NM_080675.3(SUN5):c.824C>T (p.Thr275Met)	SUN5	20	32985809	SPERMATOGENIC FAILURE 16
NM_080675.3(SUN5):c.1066C>T (p.Arg356Cys)	SUN5	20	32983868	SPERMATOGENIC FAILURE 16
NM_080675.3(SUN5):c.781G>A (p.Val261Met)	SUN5	20	32985852	SPERMATOGENIC FAILURE 16
NM_052961.3(SLC26A8):c.260G>A (p.Arg87Gln)	SLC26A8	6	36012301	Spermatogenic failure 3
NM_052961.3(SLC26A8):c.2434G>A (p.Glu812Lys)	SLC26A8	6	35951201	Spermatogenic failure 3
NM_052961.3(SLC26A8):c.2860C>T (p.Arg954Cys)	SLC26A8	6	35943953	Spermatogenic failure 3
NM_004959.4(NR5A1):c.392C>T (p.Pro131Leu)	NR5A1	9	124500568	Spermatogenic failure 8
NM_004959.4(NR5A1):c.634G>A (p.Gly212Ser)	NR5A1	9	124500326	Spermatogenic failure 8
NM_173812.4(DPY19L2):c.869G>A (p.Arg290His)	DPY19L2	12	63624124	Spermatogenic failure 9
NM_173812.4(DPY19L2):c.892C>T (p.Arg298Cys)	DPY19L2	12	63624101	Spermatogenic failure 9
NM_173812.4(DPY19L2):c.1218+1G>A	DPY19L2	12	63617303	Spermatogenic failure 9
NM_001003811.1(TEX11):c.2092G>A (p.Ala698Thr)	TEX11	X	70605421	Spermatogenic failure, X-linked, 2
NM_001003811.1(TEX11):c.450C>T (p.Ala150=)	TEX11	X	70853248	Spermatogenic failure, X-linked, 2
NM_001142446.1(ANK1):c.5286G>A (p.Trp1762Ter)	ANK1	8	41668498	Spherocytosis type 1
NM_001142446.1(ANK1):c.5620C>T (p.Arg1874Ter)	ANK1	8	41661923	Spherocytosis type 1
NM_001024858.2(SPTB):c.5266C>T (p.Arg1756Ter)	SPTB	14	64772867	Spherocytosis type 2

NM_000342.3(SLC4A1):c.-62G>A	SLC4A1	17	44262928	Spherocytosis type 4
NM_000342.3(SLC4A1):c.2312G>A (p.Gly771Asp)	SLC4A1	17	44251588	Spherocytosis type 4
NM_000342.3(SLC4A1):c.988C>T (p.Gln330Ter)	SLC4A1	17	44258512	Spherocytosis type 4
NM_000342.3(SLC4A1):c.448C>T (p.Arg150Ter)	SLC4A1	17	44260441	Spherocytosis type 4
NM_000342.3(SLC4A1):c.388G>A (p.Gly130Arg)	SLC4A1	17	44260501	Spherocytosis type 4
NM_000342.3(SLC4A1):c.268G>A (p.Glu90Lys)	SLC4A1	17	44260716	Spherocytosis type 4
NM_000342.3(SLC4A1):c.2608C>T (p.Arg870Trp)	SLC4A1	17	44251206	Spherocytosis type 4
NM_000342.3(SLC4A1):c.2279G>A (p.Arg760Gln)	SLC4A1	17	44253150	Spherocytosis type 4
NM_000119.2(EPB42):c.424G>A (p.Ala142Thr)	EPB42	15	43215191	Spherocytosis type 5
NM_000119.2(EPB42):c.929G>A (p.Arg310Gln)	EPB42	15	43208769	Spherocytosis type 5
NM_000119.2(EPB42):c.922+1G>A	EPB42	15	43209273	Spherocytosis type 5
NM_000119.2(EPB42):c.920C>T (p.Thr307Ile)	EPB42	15	43209276	Spherocytosis type 5
NM_000119.2(EPB42):c.949C>T (p.Arg317Cys)	EPB42	15	43208749	Spherocytosis type 5
NM_020475.2(ANK1):c.-153G>A	ANK1	8	41797691	Spherocytosis, type 1, autosomal recessive
NM_001142446.1(ANK1):c.1900-17G>A	ANK1	8	41708992	Spherocytosis, type 1, autosomal recessive
NM_006790.2(MYOT):c.116C>T (p.Ser39Phe)	MYOT	5	137870767	Spheroid body myopathy
NM_001042465.2(PSAP):c.650C>T (p.Thr217Ile)	PSAP	10	71828084	Sphingolipid activator protein 1 deficiency
NM_015247.2(CYLD):c.2108G>A (p.Arg703Lys)	CYLD	16	50787852	Spiegler-Brooke syndrome
NM_016213.4(TRIP4):c.760C>T (p.Arg254Ter)	TRIP4	15	64406392	Spinal muscular atrophy with congenital bone fractures 1
NM_016213.4(TRIP4):c.832C>T (p.Arg278Ter)	TRIP4	15	64409617	Spinal muscular atrophy with congenital bone fractures 1
NM_002180.2(IGHMBP2):c.1540G>A (p.Glu514Lys)	IGHMBP2	11	68934466	Spinal muscular atrophy, distal, autosomal recessive, 1;Werdnig-Hoffmann disease;not provided
NM_001039550.1(DNAJB2):c.352+1G>A	DNAJB2	2	219282062	Spinal muscular atrophy, distal, autosomal recessive, 5
NM_001039550.1(DNAJB2):c.620-1G>A	DNAJB2	2	219284631	Spinal muscular atrophy, distal, autosomal recessive, 5
NM_015250.3(BICD2):c.320C>T (p.Ser107Leu)	BICD2	9	92729157	Spinal muscular atrophy, lower extremity predominant 2, autosomal dominant
NM_001003800.1(BICD2):c.2108C>T (p.Thr703Met)	BICD2	9	92717947	Spinal muscular atrophy, lower extremity predominant 2, autosomal dominant;not provided
NM_000344.3(SMN1):c.88G>A (p.Asp30Asn)	SMN1	5	70938845	Spinal muscular atrophy, type II
NM_004977.2(KCNC3):c.1259G>A (p.Arg420His)	KCNC3	19	50323694	Spinocerebellar ataxia 13
NM_004977.2(KCNC3):c.1283C>T (p.Thr428Ile)	KCNC3	19	50323670	Spinocerebellar ataxia 13
NM_002739.4(PRKCG):c.301C>T (p.His101Tyr)	PRKCG	19	53889653	Spinocerebellar ataxia 14
NM_002739.4(PRKCG):c.383G>A (p.Gly128Asp)	PRKCG	19	53889735	Spinocerebellar ataxia 14
NM_002739.4(PRKCG):c.353G>A (p.Gly118Asp)	PRKCG	19	53889705	Spinocerebellar ataxia 14
NM_002739.4(PRKCG):c.1078G>A (p.Gly360Ser)	PRKCG	19	53898097	Spinocerebellar ataxia 14
NM_002739.4(PRKCG):c.341G>A (p.Cys114Tyr)	PRKCG	19	53889693	Spinocerebellar ataxia 14
NM_002739.4(PRKCG):c.356C>T (p.Ser119Phe)	PRKCG	19	53889708	Spinocerebellar ataxia 14
NM_002739.4(PRKCG):c.367G>A (p.Gly123Arg)	PRKCG	19	53889719	Spinocerebellar ataxia 14
NM_002739.4(PRKCG):c.368G>A (p.Gly123Glu)	PRKCG	19	53889720	Spinocerebellar ataxia 14
NM_002739.4(PRKCG):c.392G>A (p.Cys131Tyr)	PRKCG	19	53889744	Spinocerebellar ataxia 14
NM_002222.5(ITPR1):c.3176C>T (p.Pro1059Leu)	ITPR1	3	4683472	Spinocerebellar ataxia 15
NM_001168272.1(ITPR1):c.7739G>A (p.Gly2580Glu)	ITPR1	3	4815135	Spinocerebellar ataxia 15
NM_001114748.1(TMEM240):c.346C>T (p.Arg116Cys)	TMEM240	1	1535616	Spinocerebellar ataxia 21
NM_001114748.1(TMEM240):c.239C>T (p.Thr80Met)	TMEM240	1	1535723	Spinocerebellar ataxia 21
NM_001114748.1(TMEM240):c.511C>T (p.Arg171Trp)	TMEM240	1	1535370	Spinocerebellar ataxia 21
NM_020680.3(SCYL1):c.1230+1G>A	SCYL1	11	65532806	Spinocerebellar ataxia 21
NM_020680.3(SCYL1):c.1636C>T (p.Gln546Ter)	SCYL1	11	65536319	Spinocerebellar ataxia 21
NM_001114748.1(TMEM240):c.509C>T (p.Pro170Leu)	TMEM240	1	1535372	Spinocerebellar ataxia 21;not provided

NM_024411.4(PDYN):c.643C>T (p.Arg215Cys)	PDYN	20	1980445	Spinocerebellar ataxia 23	
NM_024411.4(PDYN):c.634C>T (p.Arg212Trp)	PDYN	20	1980454	Spinocerebellar ataxia 23	
NM_006796.2(AFG3L2):c.2071G>A (p.Glu691Lys)	AFG3L2	18	12337445	Spinocerebellar ataxia 28	
NM_006796.2(AFG3L2):c.2105G>A (p.Arg702Gln)	AFG3L2	18	12337411	Spinocerebellar ataxia 28	
NM_006796.2(AFG3L2):c.2011G>A (p.Gly671Arg)	AFG3L2	18	12337505	Spinocerebellar ataxia 28	
NM_006796.2(AFG3L2):c.1961C>T (p.Thr654Ile)	AFG3L2	18	12340220	Spinocerebellar ataxia 28	
NM_006796.2(AFG3L2):c.2012G>A (p.Gly671Glu)	AFG3L2	18	12337504	Spinocerebellar ataxia 28	
NM_006796.2(AFG3L2):c.2098G>A (p.Glu700Lys)	AFG3L2	18	12337418	Spinocerebellar ataxia 28	
NM_001168272.1(ITPR1):c.4639G>A (p.Val1547Met)	ITPR1	3	4706193	Spinocerebellar ataxia 29	
NM_198994.2(TGM6):c.331C>T (p.Arg111Cys)	TGM6	20	2395343	Spinocerebellar ataxia 35	
NM_001080414.3(CCDC88C):c.1391G>A (p.Arg464His)	CCDC88C	14	91321256	Spinocerebellar ataxia 40	
NM_001130698.1(TRPC3):c.2285G>A (p.Arg762His)	TRPC3	4	121903030	Spinocerebellar ataxia 41	
NM_018896.4(CACNA1G):c.5144G>A (p.Arg1715His)	CACNA1G	17	50617560	Spinocerebellar ataxia 42	
NM_000902.3(MME):c.428G>A (p.Cys143Tyr)	MME	3	155116548	Spinocerebellar ataxia 43	
NM_006946.2(SPTBN2):c.1438C>T (p.Arg480Trp)	SPTBN2	11	66707731	Spinocerebellar ataxia 5	
NM_015046.5(SETX):c.4087C>T (p.Arg1363Ter)	SETX	9	132327511	Spinocerebellar ataxia autosomal recessive 1	
NM_015046.5(SETX):c.2602C>T (p.Gln868Ter)	SETX	9	132328996	Spinocerebellar ataxia autosomal recessive 1	
NM_015046.5(SETX):c.6638C>T (p.Pro2213Leu)	SETX	9	132281483	Spinocerebellar ataxia autosomal recessive 1	
NM_015046.5(SETX):c.5929C>T (p.Leu1977Phe)	SETX	9	132296907	Spinocerebellar ataxia autosomal recessive 1	
NM_015046.5(SETX):c.6322C>T (p.Gln2108Ter)	SETX	9	132288238	Spinocerebellar ataxia autosomal recessive 1	
NM_015046.5(SETX):c.994C>T (p.Arg332Trp)	SETX	9	132331293	Spinocerebellar ataxia autosomal recessive 1;not provided	
NM_018075.4(ANO10):c.1843G>A (p.Asp615Asn)	ANO10	3	43432682	Spinocerebellar ataxia, autosomal recessive 10	
NM_018075.4(ANO10):c.337+1G>A	ANO10	3	43600383	Spinocerebellar ataxia, autosomal recessive 10	
NM_001146261.2(SYT14):c.1451G>A (p.Gly484Asp)	SYT14	1	210155872	Spinocerebellar ataxia, autosomal recessive 11	
NM_005861.3(STUB1):c.493C>T (p.Leu165Phe)	STUB1	16	681572	Spinocerebellar ataxia, autosomal recessive 16	
NM_005861.3(STUB1):c.737C>T (p.Thr246Met)	STUB1	16	682232	Spinocerebellar ataxia, autosomal recessive 16	
NM_005861.3(STUB1):c.235G>A (p.Ala79Thr)	STUB1	16	681227	Spinocerebellar ataxia, autosomal recessive 16	
NM_018294.5(CWF19L1):c.964+1G>A	CWF19L1	10	100245798	Spinocerebellar ataxia, autosomal recessive 17;not provided	
NM_015160.2(PMPCA):c.287C>T (p.Ser96Leu)	PMPCA	9	136412502	Spinocerebellar ataxia, autosomal recessive 2	
NM_015160.2(PMPCA):c.1543G>A (p.Gly515Arg)	PMPCA	9	136423229	Spinocerebellar ataxia, autosomal recessive 2	
NM_015160.2(PMPCA):c.766G>A (p.Val256Met)	PMPCA	9	136417083	Spinocerebellar ataxia, autosomal recessive 2	
NM_015160.2(PMPCA):c.64C>T (p.Arg22Trp)	PMPCA	9	136410732	Spinocerebellar ataxia, autosomal recessive 2	
NM_015160.2(PMPCA):c.554G>A (p.Arg185Gln)	PMPCA	9	136416312	Spinocerebellar ataxia, autosomal recessive 2	
NM_153816.5(SNX14):c.2596C>T (p.Gln866Ter)	SNX14	6	85513857	Spinocerebellar ataxia, autosomal recessive 20	
NM_153816.5(SNX14):c.1132C>T (p.Arg378Ter)	SNX14	6	85543737	Spinocerebellar ataxia, autosomal recessive 20	
NM_016614.2(TDP2):c.425+1G>A	TDP2	6	24638560	Spinocerebellar ataxia, autosomal recessive 23	
NM_024818.4(UBA5):c.736C>T (p.Arg246Ter)	UBA5	3	132672101	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 24	
NM_033071.3(SYNE1):c.22918C>T (p.Gln7640Ter)	SYNE1	6	152201838	Spinocerebellar ataxia, autosomal recessive 8	
NM_033071.3(SYNE1):c.22156C>T (p.Gln7386Ter)	SYNE1	6	152213737	Spinocerebellar ataxia, autosomal recessive 8	
NM_033071.3(SYNE1):c.10789C>T (p.Arg3597Ter)	SYNE1	6	152354817	Spinocerebellar ataxia, autosomal recessive 8	
NM_033071.3(SYNE1):c.24865C>T (p.Gln8289Ter)	SYNE1	6	152143733	Spinocerebellar ataxia, autosomal recessive 8	
NM_182961.3(SYNE1):c.24577C>T (p.Arg8193Ter)	SYNE1	6	152149542	Spinocerebellar ataxia, autosomal recessive 8	
NM_182961.3(SYNE1):c.8866C>T (p.Gln2956Ter)	SYNE1	6	152381149	Spinocerebellar ataxia, autosomal recessive 8	
NM_001271696.1(ABCB7):c.2044G>A (p.Gly682Ser)	ABCB7	X	75053585	Spinocerebellar ataxia, X-linked	
NM_001001344.2(ATP2B3):c.3320G>A (p.Gly1107Asp)	ATP2B3	X	153565081	Spinocerebellar ataxia, X-linked 1;not provided	

NM_003722.4(TP63):c.955C>T (p.Arg319Cys)	TP63	3	189867905	Split-hand/foot malformation 4
NM_003722.4(TP63):c.289C>T (p.Arg97Cys)	TP63	3	189738739	Split-hand/foot malformation 4
NM_003394.3(WNT10B):c.994C>T (p.Arg332Trp)	WNT10B	12	48966271	Split-hand/foot malformation 6
NM_001457.3(FLNB):c.2452C>T (p.Arg818Ter)	FLNB	3	58110138	Spondyiocarpotarsal synostosis syndrome
NM_001457.3(FLNB):c.4819C>T (p.Arg1607Ter)	FLNB	3	58136126	Spondyiocarpotarsal synostosis syndrome
NM_001457.3(FLNB):c.6010C>T (p.Arg2004Ter)	FLNB	3	58148771	Spondyiocarpotarsal synostosis syndrome
NM_001457.3(FLNB):c.1945C>T (p.Arg649Ter)	FLNB	3	58108461	Spondyiocarpotarsal synostosis syndrome
NM_001128225.2(SLC39A13):c.221G>A (p.Gly74Asp)	SLC39A13	11	47410315	Spondyiocheirodysplasia, Ehlers-Danlos syndrome-like
NM_016941.3(DLL3):c.1154G>A (p.Gly385Asp)	DLL3	19	39507099	Spondyiocostal dysostosis 1, autosomal recessive
NM_016941.3(DLL3):c.712C>T (p.Arg238Ter)	DLL3	19	39504130	Spondyiocostal dysostosis 1, autosomal recessive
NM_016941.3(DLL3):c.1511G>A (p.Gly504Asp)	DLL3	19	39507456	Spondyiocostal dysostosis 1, autosomal recessive
NM_032580.3(HES7):c.73C>T (p.Arg25Trp)	HES7	17	8123096	Spondyiocostal dysostosis 4, autosomal recessive;Spondyiocostal dysostosis 5
NM_004608.3(TBX6):c.844C>T (p.Arg282Ter)	TBX6	16	30086847	Spondyiocostal dysostosis 5
NM_004608.3(TBX6):c.434C>T (p.Pro145Leu)	TBX6	16	30089130	Spondyiocostal dysostosis 5
NM_001111035.2(ACP5):c.266C>T (p.Thr89Ile)	ACP5	19	11576839	Spondyloenchondrodysplasia with immune dysregulation
NM_001111035.2(ACP5):c.667C>T (p.Gln223Ter)	ACP5	19	11576311	Spondyloenchondrodysplasia with immune dysregulation
NM_001111035.2(ACP5):c.325G>A (p.Gly109Arg)	ACP5	19	11576780	Spondyloenchondrodysplasia with immune dysregulation
NM_001111035.2(ACP5):c.131C>T (p.Thr44Met)	ACP5	19	11577187	Spondyloenchondrodysplasia with immune dysregulation
NM_018946.3(NANS):c.448+1G>A	NANS	9	98077018	Spondyloepimetaphyseal dysplasia Genevieve type
NM_018946.3(NANS):c.452G>A (p.Arg151His)	NANS	9	98078196	Spondyloepimetaphyseal dysplasia Genevieve type
NM_018946.3(NANS):c.709C>T (p.Arg237Cys)	NANS	9	98080921	Spondyloepimetaphyseal dysplasia Genevieve type
NM_001844.4(COL2A1):c.1060G>A (p.Gly354Arg)	COL2A1	12	47989769	Spondyloepimetaphyseal dysplasia Strudwick type
NM_001844.4(COL2A1):c.1510G>A (p.Gly504Ser)	COL2A1	12	47986353	Spondyloepimetaphyseal dysplasia Strudwick type;Spondyloepiphyseal dysplasia congenita;Stickler syndrome type 1
NM_080605.3(B3GALT6):c.694C>T (p.Arg232Cys)	B3GALT6	1	1232972	Spondyloepimetaphyseal dysplasia with joint laxity
NM_080605.3(B3GALT6):c.466G>A (p.Asp156Asn)	B3GALT6	1	1232744	Spondyloepimetaphyseal dysplasia with joint laxity
NM_080605.3(B3GALT6):c.200C>T (p.Pro67Leu)	B3GALT6	1	1232478	Spondyloepimetaphyseal dysplasia with joint laxity
NM_080605.3(B3GALT6):c.649G>A (p.Gly217Ser)	B3GALT6	1	1232927	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with fractures
NM_007317.2(KIF22):c.442C>T (p.Pro148Ser)	KIF22	16	29798640	Spondyloepimetaphyseal dysplasia with multiple dislocations
NM_007317.2(KIF22):c.443C>T (p.Pro148Leu)	KIF22	16	29798641	Spondyloepimetaphyseal dysplasia with multiple dislocations;not provided
NM_007317.2(KIF22):c.446G>A (p.Arg149Gln)	KIF22	16	29798644	Spondyloepimetaphyseal dysplasia with multiple dislocations;not provided
NM_013227.3(ACAN):c.7141G>A (p.Asp2381Asn)	ACAN	15	88872038	Spondyloepimetaphyseal dysplasia, Aggrecan type
NM_001015880.1(PAPSS2):c.1000C>T (p.Arg334Ter)	PAPSS2	10	87727403	Spondyloepimetaphyseal dysplasia, pakistani type
NM_001844.4(COL2A1):c.3301G>A (p.Gly1101Arg)	COL2A1	12	47977128	Spondyloepiphyseal dysplasia congenita
NM_001844.4(COL2A1):c.1636G>A (p.Gly546Ser)	COL2A1	12	47985772	Spondyloepiphyseal dysplasia congenita
NM_001844.4(COL2A1):c.3589G>A (p.Gly1197Ser)	COL2A1	12	47975971	Spondyloepiphyseal dysplasia congenita;not provided
NM_001844.4(COL2A1):c.2965C>T (p.Arg989Cys)	COL2A1	12	47978329	Spondyloepiphyseal dysplasia congenita;not provided
NM_001844.4(COL2A1):c.4316C>T (p.Thr1439Met)	COL2A1	12	47974090	Spondyloepiphyseal dysplasia congenita;Spondyloperipheral dysplasia
NM_001011658.3(TRAPPC2):c.934-5G>A	TRAPPC2	X	13719866	Spondyloepiphyseal dysplasia tarda
NM_001011658.3(TRAPPC2):c.391C>T (p.Gln131Ter)	TRAPPC2	X	13714439	Spondyloepiphyseal dysplasia tarda
NM_004273.4(CHST3):c.911G>A (p.Arg304Gln)	CHST3	10	72007942	Spondyloepiphyseal dysplasia with congenital joint dislocations
NM_004273.4(CHST3):c.1114G>A (p.Glu372Lys)	CHST3	10	72008145	Spondyloepiphyseal dysplasia with congenital joint dislocations
NM_004273.4(CHST3):c.664C>T (p.Arg222Trp)	CHST3	10	72007695	Spondyloepiphyseal dysplasia with congenital joint dislocations
NM_004273.4(CHST3):c.422C>T (p.Thr141Met)	CHST3	10	72007453	Spondyloepiphyseal dysplasia with congenital joint dislocations
NM_004273.4(CHST3):c.481C>T (p.Leu161Phe)	CHST3	10	72007512	Spondyloepiphyseal dysplasia with congenital joint dislocations

NM_004273.4(CHST3):c.988C>T (p.Gln330Ter)	CHST3	10	72008019	Spondyloepiphyseal dysplasia with congenital joint dislocations
NM_004273.4(CHST3):c.1063G>A (p.Gly355Arg)	CHST3	10	72008094	Spondyloepiphyseal dysplasia with congenital joint dislocations
NM_001844.4(COL2A1):c.619G>A (p.Gly207Arg)	COL2A1	12	47995910	Spondyloepiphyseal dysplasia, stanescu type
NM_001014796.1(DDR2):c.2254C>T (p.Arg752Cys)	DDR2	1	162776341	Spondylometaepiphyseal dysplasia short limb-hand type
NM_001014796.1(DDR2):c.2138C>T (p.Thr713Ile)	DDR2	1	162776225	Spondylometaepiphyseal dysplasia short limb-hand type
NM_001014796.1(DDR2):c.337G>A (p.Glu113Lys)	DDR2	1	162754775	Spondylometaepiphyseal dysplasia short limb-hand type
NM_002085.4(GPX4):c.476+5G>A	GPX4	19	1105814	Spondylometaphyseal dysplasia Sedaghatian type
NM_005017.3(PCYT1A):c.296C>T (p.Ala99Val)	PCYT1A	3	196248245	Spondylometaphyseal dysplasia with cone-rod dystrophy
NM_005017.3(PCYT1A):c.295G>A (p.Ala99Thr)	PCYT1A	3	196248246	Spondylometaphyseal dysplasia with cone-rod dystrophy
NM_005017.3(PCYT1A):c.847C>T (p.Arg283Ter)	PCYT1A	3	196239597	Spondylometaphyseal dysplasia with cone-rod dystrophy
NM_005017.3(PCYT1A):c.385G>A (p.Glu129Lys)	PCYT1A	3	196247468	Spondylometaphyseal dysplasia with cone-rod dystrophy
NM_005450.4(NOG):c.328C>T (p.Gln110Ter)	NOG	17	56594551	Stapes ankylosis with broad thumb and toes
NM_000350.2(ABCA4):c.3083C>T (p.Ala1028Val)	ABCA4	1	94043443	Stargardt disease 1
NM_000350.2(ABCA4):c.4429C>T (p.Gln1477Ter)	ABCA4	1	94029555	Stargardt disease 1
NM_000350.2(ABCA4):c.6647C>T (p.Ala2216Val)	ABCA4	1	93997943	Stargardt disease 1
NM_000350.2(ABCA4):c.5312+1G>A	ABCA4	1	94015738	Stargardt disease 1
NM_000350.2(ABCA4):c.5189G>A (p.Trp1730Ter)	ABCA4	1	94019589	Stargardt disease 1
NM_000350.2(ABCA4):c.4352+1G>A	ABCA4	1	94030427	Stargardt disease 1
NM_000350.2(ABCA4):c.4253+5G>A	ABCA4	1	94030991	Stargardt disease 1
NM_000350.2(ABCA4):c.3871C>T (p.Gln1291Ter)	ABCA4	1	94032035	Stargardt disease 1
NM_000350.2(ABCA4):c.3813G>A (p.Glu1271=)	ABCA4	1	94037145	Stargardt disease 1
NM_000350.2(ABCA4):c.1293G>A (p.Trp431Ter)	ABCA4	1	94078653	Stargardt disease 1
NM_000350.2(ABCA4):c.206G>A (p.Trp69Ter)	ABCA4	1	94111534	Stargardt disease 1
NM_000350.2(ABCA4):c.2565G>A (p.Trp855Ter)	ABCA4	1	94055133	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.3106G>A (p.Glu1036Lys)	ABCA4	1	94043420	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.52C>T (p.Arg18Trp)	ABCA4	1	94120994	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.3364G>A (p.Glu1122Lys)	ABCA4	1	94041367	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.3322C>T (p.Arg1108Cys)	ABCA4	1	94042767	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.122G>A (p.Trp41Ter)	ABCA4	1	94113011	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.1804C>T (p.Arg602Trp)	ABCA4	1	94062710	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.1903C>T (p.Gln635Ter)	ABCA4	1	94062611	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.1937+1G>A	ABCA4	1	94062576	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.194G>A (p.Gly65Glu)	ABCA4	1	94111546	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.2564G>A (p.Trp855Ter)	ABCA4	1	94055134	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.3085C>T (p.Gln1029Ter)	ABCA4	1	94043441	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.4195G>A (p.Glu1399Lys)	ABCA4	1	94031054	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.4234C>T (p.Gln1412Ter)	ABCA4	1	94031015	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.4457C>T (p.Pro1486Leu)	ABCA4	1	94029527	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.454C>T (p.Arg152Ter)	ABCA4	1	94103131	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.4594G>A (p.Asp1532Asn)	ABCA4	1	94024994	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.45G>A (p.Trp15Ter)	ABCA4	1	94121001	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.4610C>T (p.Thr1537Met)	ABCA4	1	94024978	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.4919G>A (p.Arg1640Gln)	ABCA4	1	94021339	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.5196+1G>A	ABCA4	1	94019581	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.6089G>A (p.Arg2030Gln)	ABCA4	1	94005499	Stargardt disease 1;not provided

NM_000350.2(ABCA4):c.6112C>T (p.Arg2038Trp)	ABCA4	1	94005476	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.6118C>T (p.Arg2040Ter)	ABCA4	1	94005470	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.6316C>T (p.Arg2106Cys)	ABCA4	1	94001072	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.6342G>A (p.Val2114=)	ABCA4	1	94001046	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.6658C>T (p.Gln2220Ter)	ABCA4	1	93997932	Stargardt disease 1;not provided
NM_000350.2(ABCA4):c.3056C>T (p.Thr1019Met)	ABCA4	1	94043470	Stargardt disease 1;not provided
NM_001844.4(COL2A1):c.2794C>T (p.Arg932Ter)	COL2A1	12	47978698	Stickler syndrome type 1
NM_001844.4(COL2A1):c.625C>T (p.Arg209Ter)	COL2A1	12	47995904	Stickler syndrome type 1
NM_001844.4(COL2A1):c.762+1G>A	COL2A1	12	47995254	Stickler syndrome type 1
NM_001844.4(COL2A1):c.2530C>T (p.Gln844Ter)	COL2A1	12	47980649	Stickler syndrome type 1
NM_001844.4(COL2A1):c.3574C>T (p.Arg1192Ter)	COL2A1	12	47975986	Stickler syndrome type 1
NM_001844.4(COL2A1):c.2353C>T (p.Arg785Ter)	COL2A1	12	47982109	Stickler syndrome type 1
NM_001844.4(COL2A1):c.1693C>T (p.Arg565Cys)	COL2A1	12	47985575	Stickler syndrome type 1;not provided
NM_001844.4(COL2A1):c.3106C>T (p.Arg1036Ter)	COL2A1	12	47978015	Stickler syndrome type 1;not provided
NM_001844.4(COL2A1):c.1999C>T (p.Leu667Phe)	COL2A1	12	47983435	Stickler syndrome type 1;Stickler syndrome, type I, nonsyndromic ocular
NM_080680.2(COL11A2):c.4392+1G>A	COL11A2	6	33166512	Stickler syndrome, type 3
NM_080680.2(COL11A2):c.4135C>T (p.Arg1379Ter)	COL11A2	6	33167305	Stickler syndrome, type 3
NM_001851.4(COL9A1):c.883C>T (p.Arg295Ter)	COL9A1	6	70281033	Stickler syndrome, type 4
NM_001851.4(COL9A1):c.1519C>T (p.Arg507Ter)	COL9A1	6	70255375	Stickler syndrome, type 4
NM_001844.4(COL2A1):c.800G>A (p.Gly267Asp)	COL2A1	12	47994440	Stickler syndrome, type I, nonsyndromic ocular
NM_001844.4(COL2A1):c.141G>A (p.Trp47Ter)	COL2A1	12	48000070	Stickler syndrome, type I, nonsyndromic ocular
NM_001844.4(COL2A1):c.170G>A (p.Cys57Tyr)	COL2A1	12	48000041	Stickler syndrome, type I, nonsyndromic ocular
NM_198282.3(TMEM173):c.463G>A (p.Val155Met)	TMEM173	5	139480847	Sting-associated vasculopathy, infantile-onset;not provided
NM_020717.3(SHROOM4):c.3266C>T (p.Ser1089Leu)	SHROOM4	X	50607876	Stocco dos Santos syndrome
NM_006516.2(SLC2A1):c.857G>A (p.Gly286Asp)	SLC2A1	1	42929603	Stomatin-deficient cryohydrocytosis with neurologic defects
NM_003156.3(STIM1):c.910C>T (p.Arg304Trp)	STIM1	11	4074620	Stormorken syndrome
NM_021734.4(SLC25A19):c.373G>A (p.Gly125Ser)	SLC25A19	17	75283509	Striatal necrosis, bilateral, and progressive polyneuropathy
NM_018052.3(VAC14):c.1528+1G>A	VAC14	16	70744422	Striatonigral degeneration, childhood-onset
NM_018052.3(VAC14):c.1748C>T (p.Ser583Leu)	VAC14	16	70698725	Striatonigral degeneration, childhood-onset
NM_016343.3(CENPF):c.8692C>T (p.Arg2898Ter)	CENPF	1	214657139	Stromme syndrome
NM_016343.3(CENPF):c.9280C>T (p.Arg3094Ter)	CENPF	1	214663729	Stromme syndrome
NM_002310.5(LIFR):c.1789C>T (p.Arg597Ter)	LIFR	5	38496478	Stuve-Wiedemann syndrome
NM_002310.5(LIFR):c.2074C>T (p.Arg692Ter)	LIFR	5	38490283	Stuve-Wiedemann syndrome
NM_178152.2(DCX):c.233G>A (p.Arg78His)	DCX	X	111410166	Subcortical laminar heterotopia, X-linked
NM_170740.1(ALDH5A1):c.1273C>T (p.Arg425Ter)	ALDH5A1	6	24528057	Succinate-semialdehyde dehydrogenase deficiency
NM_001080.3(ALDH5A1):c.1226G>A (p.Gly409Asp)	ALDH5A1	6	24528049	Succinate-semialdehyde dehydrogenase deficiency
NM_170740.1(ALDH5A1):c.612G>A (p.Trp204Ter)	ALDH5A1	6	24504871	Succinate-semialdehyde dehydrogenase deficiency;not provided
NM_000436.3(OXCT1):c.971G>A (p.Gly324Glu)	OXCT1	5	41803148	Succinyl-CoA acetoacetate transferase deficiency
NM_000436.3(OXCT1):c.656G>A (p.Gly219Glu)	OXCT1	5	41842690	Succinyl-CoA acetoacetate transferase deficiency
NM_000436.3(OXCT1):c.661G>A (p.Val221Met)	OXCT1	5	41842685	Succinyl-CoA acetoacetate transferase deficiency
NM_001041.3(SI):c.3686G>A (p.Cys1229Tyr)	SI	3	165017621	Sucrase-isomaltase deficiency
NM_001041.3(SI):c.3218G>A (p.Gly1073Asp)	SI	3	165021265	Sucrase-isomaltase deficiency
NM_176869.2(PPA2):c.683C>T (p.Pro228Leu)	PPA2	4	105399137	SUDDEN CARDIAC FAILURE, ALCOHOL-INDUCED
NM_176869.2(PPA2):c.514G>A (p.Glu172Lys)	PPA2	4	105437964	SUDDEN CARDIAC FAILURE, ALCOHOL-INDUCED;SUDDEN CARDIAC FAILURE, INFANTILE
NM_176869.2(PPA2):c.182C>T (p.Ser61Phe)	PPA2	4	105456721	SUDDEN CARDIAC FAILURE, INFANTILE

NM_176869.2(PPA2):c.500C>T (p.Pro167Leu)	PPA2	4	105437978	SUDDEN CARDIAC FAILURE, INFANTILE
NM_000456.2(SUOX):c.650G>A (p.Arg217Gln)	SUOX	12	56004039	Sulfite oxidase deficiency, isolated
NM_000456.2(SUOX):c.1589G>A (p.Gly530Asp)	SUOX	12	56004978	Sulfite oxidase deficiency, isolated
NM_000501.3(ELN):c.1097-1G>A	ELN	7	74054715	Supravalvar aortic stenosis
NM_000501.3(ELN):c.1918+1G>A	ELN	7	74063370	Supravalvar aortic stenosis
NM_000501.3(ELN):c.1324C>T (p.Gln442Ter)	ELN	7	74056680	Supravalvar aortic stenosis;not provided
NM_000501.3(ELN):c.1621C>T (p.Arg541Ter)	ELN	7	74060184	Supravalvar aortic stenosis;not provided
NM_000501.3(ELN):c.1150+1G>A	ELN	7	74054770	Supravalvar aortic stenosis;not provided
NM_198843.2(SFTPB):c.742C>T (p.Arg248Cys)	SFTPB	2	85663814	Surfactant metabolism dysfunction, pulmonary, 1
NM_003018.3(SFTPC):c.196G>A (p.Glu66Lys)	SFTPC	8	22162727	Surfactant metabolism dysfunction, pulmonary, 2
NM_001089.2(ABCA3):c.3426G>A (p.Trp1142Ter)	ABCA3	16	2285499	Surfactant metabolism dysfunction, pulmonary, 3
NM_001161529.1(CSF2RA):c.586G>A (p.Gly196Arg)	CSF2RA	X	1290449	Surfactant metabolism dysfunction, pulmonary, 4
NM_001161529.1(CSF2RA):c.586G>A (p.Gly196Arg)	CSF2RA	Y	1290449	Surfactant metabolism dysfunction, pulmonary, 4
NM_000395.2(CSF2RB):c.812C>T (p.Ser271Leu)	CSF2RB	22	36930468	Surfactant metabolism dysfunction, pulmonary, 5
NM_001025107.2(ADAR):c.535C>T (p.Arg179Ter)	ADAR	1	154601222	Symmetrical dyschromatosis of extremities
NM_001025107.2(ADAR):c.1192C>T (p.Gln398Ter)	ADAR	1	154597125	Symmetrical dyschromatosis of extremities
NM_000557.4(GDF5):c.1471G>A (p.Glu491Lys)	GDF5	20	35433944	Symphalangism, proximal, 1b
NM_002334.3(LRP4):c.1585G>A (p.Asp529Asn)	LRP4	11	46893085	Syndactyly Cenani Lenz type
NM_002334.3(LRP4):c.409G>A (p.Asp137Asn)	LRP4	11	46899884	Syndactyly Cenani Lenz type
NM_002334.3(LRP4):c.479G>A (p.Cys160Tyr)	LRP4	11	46899455	Syndactyly Cenani Lenz type
NM_002334.3(LRP4):c.1345G>A (p.Asp449Asn)	LRP4	11	46894784	Syndactyly Cenani Lenz type
NM_000165.4(GJA1):c.427G>A (p.Gly143Ser)	GJA1	6	121447274	Syndactyly type 3
NM_003336.3(UBE2A):c.382C>T (p.Gln128Ter)	UBE2A	X	119583178	Syndromic mental retardation, Nascimento type, X-linked
NM_003336.3(UBE2A):c.32G>A (p.Arg11Gln)	UBE2A	X	119574743	Syndromic mental retardation, Nascimento type, X-linked
NM_003588.3(CUL4B):c.1714C>T (p.Arg572Cys)	CUL4B	X	120539349	Syndromic X-linked mental retardation, Cabezas type
NM_003588.3(CUL4B):c.149C>T (p.Pro50Leu)	CUL4B	X	120560544	Syndromic X-linked mental retardation, Cabezas type
NM_000523.3(HOXD13):c.916C>T (p.Arg306Trp)	HOXD13	2	176094614	Synpolydactyly 1
NM_000523.3(HOXD13):c.742C>T (p.Gln248Ter)	HOXD13	2	176093632	Synpolydactyly 1
NM_000523.3(HOXD13):c.917G>A (p.Arg306Gln)	HOXD13	2	176094615	Synpolydactyly 1
NM_000523.3(HOXD13):c.781+1G>A	HOXD13	2	176093672	Synpolydactyly 1
NM_001098629.2(IRF5):c.*555G>A	IRF5	7	128949373	Systemic lupus erythematosus 10
NM_002653.4(PITX1):c.388G>A (p.Glu130Lys)	PITX1	5	135031290	Talipes equinovarus
NM_005502.3(ABCA1):c.2810C>T (p.Ala937Val)	ABCA1	9	104822514	Tangier disease
NM_005502.3(ABCA1):c.6241C>T (p.Arg2081Trp)	ABCA1	9	104786940	Tangier disease
NM_005502.3(ABCA1):c.4517C>T (p.Ser1506Leu)	ABCA1	9	104804668	Tangier disease
NM_005502.3(ABCA1):c.3865G>A (p.Asp1289Asn)	ABCA1	9	104814154	Tangier disease
NM_000039.2(APOA1):c.67C>T (p.Gln23Ter)	APOA1	11	116837134	Tangier disease
NM_000593.5(TAP1):c.2156G>A (p.Arg719Gln)	TAP1	6	32847132	TAP1 deficiency, somatic
NM_005676.4(RBM10):c.1235G>A (p.Trp412Ter)	RBM10	X	47180493	TARP syndrome
NM_005676.4(RBM10):c.448C>T (p.Gln150Ter)	RBM10	X	47173143	TARP syndrome
NM_000520.5(HEXA):c.1444G>A (p.Glu482Lys)	HEXA	15	72345528	Tay-Sachs disease
NM_000520.5(HEXA):c.1177C>T (p.Arg393Ter)	HEXA	15	72346680	Tay-Sachs disease
NM_000520.5(HEXA):c.629C>T (p.Ser210Phe)	HEXA	15	72351176	Tay-Sachs disease
NM_000520.5(HEXA):c.78G>A (p.Trp26Ter)	HEXA	15	72375895	Tay-Sachs disease
NM_000520.5(HEXA):c.508C>T (p.Arg170Trp)	HEXA	15	72353130	Tay-Sachs disease

NM_000520.5(HEXA):c.1360G>A (p.Gly454Ser)	HEXA	15	72346296	Tay-Sachs disease
NM_000520.5(HEXA):c.1176G>A (p.Trp392Ter)	HEXA	15	72346681	Tay-Sachs disease
NM_000520.5(HEXA):c.805+1G>A	HEXA	15	72350517	Tay-Sachs disease
NM_000520.5(HEXA):c.173G>A (p.Cys58Tyr)	HEXA	15	72375800	Tay-Sachs disease
NM_000520.5(HEXA):c.1305C>T (p.Tyr435=)	HEXA	15	72346552	Tay-Sachs disease
NM_000520.5(HEXA):c.340G>A (p.Glu114Lys)	HEXA	15	72356531	Tay-Sachs disease
NM_000520.5(HEXA):c.964G>A (p.Asp322Asn)	HEXA	15	72349101	Tay-Sachs disease
NM_000520.5(HEXA):c.1421G>A (p.Trp474Ter)	HEXA	15	72346235	Tay-Sachs disease
NM_000520.5(HEXA):c.709C>T (p.Gln237Ter)	HEXA	15	72350614	Tay-Sachs disease
NM_000520.5(HEXA):c.1454G>A (p.Trp485Ter)	HEXA	15	72345518	Tay-Sachs disease
NM_000520.5(HEXA):c.1432G>A (p.Gly478Arg)	HEXA	15	72345540	Tay-Sachs disease
NM_000520.5(HEXA):c.316C>T (p.Gln106Ter)	HEXA	15	72356555	Tay-Sachs disease
NM_000520.5(HEXA):c.749G>A (p.Gly250Asp)	HEXA	15	72350574	Tay-sachs disease, juvenile
NM_000520.5(HEXA):c.509G>A (p.Arg170Gln)	HEXA	15	72353129	Tay-Sachs disease;not provided
NM_000520.5(HEXA):c.409C>T (p.Arg137Ter)	HEXA	15	72355562	Tay-Sachs disease;not provided
NM_000520.5(HEXA):c.1073+1G>A	HEXA	15	72348047	Tay-Sachs disease;not provided
NM_000520.5(HEXA):c.672+1G>A	HEXA	15	72351132	Tay-Sachs disease;not provided
NM_000520.5(HEXA):c.1528C>T (p.Arg510Ter)	HEXA	15	72344139	Tay-Sachs disease;not provided
NM_000520.5(HEXA):c.1330+1G>A	HEXA	15	72346526	Tay-Sachs disease;not provided
NM_000520.5(HEXA):c.459+5G>A	HEXA	15	72353686	Tay-Sachs disease;not provided
NM_000520.5(HEXA):c.532C>T (p.Arg178Cys)	HEXA	15	72353106	Tay-Sachs disease;Tay-Sachs disease, B1 variant
NM_000520.5(HEXA):c.533G>A (p.Arg178His)	HEXA	15	72353105	Tay-Sachs disease;Tay-Sachs disease, B1 variant;not provided
NM_138761.3(BAX):c.199G>A (p.Gly67Arg)	BAX	19	48955799	T-cell acute lymphoblastic leukemia
NM_003593.2(FOXN1):c.763C>T (p.Arg255Ter)	FOXN1	17	28529157	T-cell immunodeficiency, congenital alopecia and nail dystrophy
NM_006282.3(STK4):c.349C>T (p.Arg117Ter)	STK4	20	44981932	T-cell immunodeficiency, recurrent infections, and autoimmunity with or without cardiac malformations
NG_001332.2:g.1074699G>A	TRAC	14	22550664	T-cell receptor alpha/beta deficiency
NM_016204.3(GDF2):c.254C>T (p.Pro85Leu)	GDF2	10	47322922	Telangiectasia, hereditary hemorrhagic, type 5
NM_016204.3(GDF2):c.997C>T (p.Arg333Trp)	GDF2	10	47325491	Telangiectasia, hereditary hemorrhagic, type 5
NM_172362.2(KCNH1):c.1546C>T (p.Leu516Phe)	KCNH1	1	210804083	Temple-Baraitser syndrome
NM_014918.4(CHSY1):c.205C>T (p.Gln69Ter)	CHSY1	15	101251252	Temtamy preaxial brachydactyly syndrome
NM_138425.3(C12orf57):c.184C>T (p.Gln62Ter)	C12orf57	12	6944607	Temtamy syndrome;not provided
NM_017831.3(RNF125):c.336G>A (p.Met112Ile)	RNF125	18	32042196	Tenorio syndrome
NM_017831.3(RNF125):c.488C>T (p.Ser163Leu)	RNF125	18	32045716	Tenorio syndrome
NM_017831.3(RNF125):c.520C>T (p.Arg174Cys)	RNF125	18	32065917	Tenorio syndrome
NM_001110556.1(FLNA):c.5217G>A (p.Thr1739=)	FLNA	X	154354825	Terminal osseous dysplasia
NM_002052.4(GATA4):c.661G>A (p.Gly221Arg)	GATA4	8	11748963	Testicular anomalies with or without congenital heart disease
NM_000197.1(HSD17B3):c.695C>T (p.Ser232Leu)	HSD17B3	9	96240885	Testosterone 17-beta-dehydrogenase deficiency
NM_000197.1(HSD17B3):c.608C>T (p.Ala203Val)	HSD17B3	9	96244393	Testosterone 17-beta-dehydrogenase deficiency
NM_000197.1(HSD17B3):c.607-1G>A	HSD17B3	9	96244395	Testosterone 17-beta-dehydrogenase deficiency
NM_000197.1(HSD17B3):c.166G>A (p.Ala56Thr)	HSD17B3	9	96298451	Testosterone 17-beta-dehydrogenase deficiency
NM_000197.1(HSD17B3):c.803G>A (p.Cys268Tyr)	HSD17B3	9	96240777	Testosterone 17-beta-dehydrogenase deficiency
NM_000197.1(HSD17B3):c.239G>A (p.Arg80Gln)	HSD17B3	9	96254906	Testosterone 17-beta-dehydrogenase deficiency;not provided
NM_030753.4(WNT3):c.247C>T (p.Gln83Ter)	WNT3	17	46773743	Tetraamelia, autosomal recessive
NM_000214.2(JAG1):c.821G>A (p.Gly274Asp)	JAG1	20	10652533	Tetralogy of Fallot

NM_004387.3(NKX2-5):c.646C>T (p.Arg216Cys)	NKX2-5	5	173232898	Tetralogy of Fallot
NM_004387.3(NKX2-5):c.656C>T (p.Ala219Val)	NKX2-5	5	173232888	Tetralogy of Fallot
NM_017617.4(NOTCH1):c.578G>A (p.Gly193Asp)	NOTCH1	9	136523014	Tetralogy of Fallot
NM_000358.2(TGFBI):c.1664G>A (p.Arg555Gln)	TGFBI	5	136056781	Thiel-Behnke corneal dystrophy
NM_003242.5(TGFBR2):c.1178G>A (p.Cys393Tyr)	TGFBR2	3	30672361	Thoracic aortic aneurysm and aortic dissection
NM_001613.2(ACTA2):c.940C>T (p.Arg314Ter)	ACTA2	10	88938111	Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.8488C>T (p.Gln2830Ter)	FBN1	15	48411118	Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.7939C>T (p.Gln2647Ter)	FBN1	15	48415648	Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.7792C>T (p.Gln2598Ter)	FBN1	15	48420714	Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.6884G>A (p.Cys2295Tyr)	FBN1	15	48428459	Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.1807C>T (p.Gln603Ter)	FBN1	15	48508612	Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.526C>T (p.Gln176Ter)	FBN1	15	48596295	Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.439C>T (p.Gln147Ter)	FBN1	15	48600142	Thoracic aortic aneurysm and aortic dissection
NM_000138.4(FBN1):c.7C>T (p.Arg3Ter)	FBN1	15	48644763	Thoracic aortic aneurysm and aortic dissection
NM_004612.3(TGFBR1):c.469C>T (p.Arg157Ter)	TGFBR1	9	99132634	Thoracic aortic aneurysm and aortic dissection
NM_001613.2(ACTA2):c.635G>A (p.Arg212Gln)	ACTA2	10	88939680	Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.6169C>T (p.Arg2057Ter)	FBN1	15	48437912	Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.4930C>T (p.Arg1644Ter)	FBN1	15	48465580	Thoracic aortic aneurysm and aortic dissection;not provided
NM_000138.4(FBN1):c.1693C>T (p.Arg565Ter)	FBN1	15	48510065	Thoracic aortic aneurysm and aortic dissection;not provided
NM_003242.5(TGFBR2):c.1489C>T (p.Arg497Ter)	TGFBR2	3	30688476	Thoracic aortic aneurysm and aortic dissection;not provided
NM_005902.3(SMAD3):c.277C>T (p.Arg93Ter)	SMAD3	15	67164965	Thoracic aortic aneurysm and aortic dissection;not provided
NM_014780.4(CUL7):c.4333C>T (p.Arg1445Ter)	CUL7	6	43038949	Three M syndrome 1
NM_014780.4(CUL7):c.2164C>T (p.Arg722Ter)	CUL7	6	43048153	Three M syndrome 1
NM_014780.4(CUL7):c.4318C>T (p.Arg1440Ter)	CUL7	6	43038964	Three M syndrome 1
NM_015311.2(OBSL1):c.1465C>T (p.Arg489Ter)	OBSL1	2	219567787	Three M syndrome 2
NM_015311.2(OBSL1):c.3955C>T (p.Gln1319Ter)	OBSL1	2	219557454	Three M syndrome 2
NM_004972.3(JAK2):c.1849G>A (p.Val617Ile)	JAK2	9	5073770	Thrombocythemia 3
NM_014915.2(ANKRD26):c.-134G>A	ANKRD26	10	27100460	Thrombocytopenia 2
NM_018947.5(CYCS):c.124G>A (p.Gly42Ser)	CYCS	7	25123996	Thrombocytopenia 4
NM_198291.2(SRC):c.1579G>A (p.Glu527Lys)	SRC	20	37403347	Thrombocytopenia 6
NM_002049.3(GATA1):c.647G>A (p.Arg216Gln)	GATA1	X	48792371	Thrombocytopenia, platelet dysfunction, hemolysis, and imbalanced globin synthesis
NM_002049.3(GATA1):c.646C>T (p.Arg216Trp)	GATA1	X	48792370	Thrombocytopenia, platelet dysfunction, hemolysis, and imbalanced globin synthesis
NM_000377.2(WAS):c.167C>T (p.Ala56Val)	WAS	X	48684317	Thrombocytopenia, X-linked
NM_000377.2(WAS):c.134C>T (p.Thr45Met)	WAS	X	48684284	Thrombocytopenia, X-linked
NM_000312.3(PROC):c.1042C>T (p.Arg348Ter)	PROC	2	127428602	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant
NM_000312.3(PROC):c.658C>T (p.Arg220Trp)	PROC	2	127426207	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant
NM_000312.3(PROC):c.659G>A (p.Arg220Gln)	PROC	2	127426208	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant
NM_000312.3(PROC):c.814C>T (p.Arg272Cys)	PROC	2	127428374	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant
NM_000312.3(PROC):c.1015G>A (p.Val339Met)	PROC	2	127428575	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant
NM_000312.3(PROC):c.935C>T (p.Ser312Leu)	PROC	2	127428495	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant
NM_000312.3(PROC):c.1201G>A (p.Asp401Asn)	PROC	2	127428761	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant
NM_000312.3(PROC):c.169C>T (p.Arg57Trp)	PROC	2	127421381	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant
NM_000312.3(PROC):c.631C>T (p.Arg211Trp)	PROC	2	127426180	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant;Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive

NM_000312.3(PROC):c.1000G>A (p.Gly334Ser)	PROC	2	127428560	Thrombophilia, hereditary, due to protein C deficiency, autosomal dominant;Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive
NM_000312.3(PROC):c.902C>T (p.Ala301Val)	PROC	2	127428462	Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive
NM_000312.3(PROC):c.629C>T (p.Pro210Leu)	PROC	2	127426178	Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive
NM_000312.3(PROC):c.925G>A (p.Ala309Thr)	PROC	2	127428485	Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive
NM_000312.3(PROC):c.1027G>A (p.Gly343Ser)	PROC	2	127428587	Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive
NM_000312.3(PROC):c.226G>A (p.Val76Met)	PROC	2	127421438	Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive
NM_000312.3(PROC):c.866C>T (p.Pro289Leu)	PROC	2	127428426	Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive
NM_000312.3(PROC):c.793C>T (p.Leu265Phe)	PROC	2	127427219	Thrombophilia, hereditary, due to protein C deficiency, autosomal recessive
NM_000412.4(HRG):c.308G>A (p.Gly103Glu)	HRG	3	186669945	Thrombophilia, histidine-rich glycoprotein-related
NM_000222.2(KIT):c.1468G>A (p.Glu490Lys)	KIT	4	54725978	Thymoma
NM_013953.3(PAX8):c.322C>T (p.Arg108Ter)	PAX8	2	113244494	Thyroid agenesis
NM_013953.3(PAX8):c.92G>A (p.Arg31His)	PAX8	2	113246853	Thyroid agenesis
NM_013953.3(PAX8):c.170G>A (p.Cys57Tyr)	PAX8	2	113246775	Thyroid agenesis
NM_013953.3(PAX8):c.143C>T (p.Ser48Phe)	PAX8	2	113246802	Thyroid agenesis
NM_004897.4(MINPP1):c.122C>T (p.Ser41Leu)	MINPP1	10	87505037	Thyroid cancer, follicular
NM_020762.3(SRGAP1):c.823G>A (p.Ala275Thr)	SRGAP1	12	64062938	Thyroid cancer, follicular
NM_020762.3(SRGAP1):c.1849C>T (p.Arg617Cys)	SRGAP1	12	64108967	Thyroid cancer, follicular
NM_000453.2(SLC5A5):c.1628G>A (p.Gly543Glu)	SLC5A5	19	17888432	Thyroid dyshormonogenesis 1
NM_000453.2(SLC5A5):c.1183G>A (p.Gly395Arg)	SLC5A5	19	17882160	Thyroid dyshormonogenesis 1
NM_014080.4(DUOX2):c.1300C>T (p.Arg434Ter)	DUOX2	15	45108887	Thyroid dyshormonogenesis 6
NM_014080.4(DUOX2):c.2056C>T (p.Gln686Ter)	DUOX2	15	45106217	Thyroid dyshormonogenesis 6
NM_014080.4(DUOX2):c.2524C>T (p.Arg842Ter)	DUOX2	15	45104176	Thyroid dyshormonogenesis 6
NM_014080.4(DUOX2):c.1126C>T (p.Arg376Trp)	DUOX2	15	45109895	Thyroid dyshormonogenesis 6
NM_024077.4(SECISBP2):c.1619G>A (p.Arg540Gln)	SECISBP2	9	89348095	Thyroid hormone metabolism, abnormal
NM_024077.4(SECISBP2):c.1212+29G>A	SECISBP2	9	89338609	Thyroid hormone metabolism, abnormal
NM_001128177.1(THRB):c.949G>A (p.Ala317Thr)	THRB	3	24127694	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.1040G>A (p.Gly347Glu)	THRB	3	24127603	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.1033G>A (p.Gly345Ser)	THRB	3	24127610	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.959G>A (p.Arg320His)	THRB	3	24127684	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.700G>A (p.Ala234Thr)	THRB	3	24143539	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.958C>T (p.Arg320Cys)	THRB	3	24127685	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.1313G>A (p.Arg438His)	THRB	3	24122957	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.728G>A (p.Arg243Gln)	THRB	3	24143511	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.727C>T (p.Arg243Trp)	THRB	3	24143512	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.1148G>A (p.Arg383His)	THRB	3	24123122	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.67C>T (p.Arg23Ter)	THRB	3	24190290	Thyroid hormone resistance, generalized, autosomal dominant
NM_001128177.1(THRB):c.994G>A (p.Gly332Arg)	THRB	3	24127649	Thyroid hormone resistance, generalized, autosomal dominant;not provided
NM_001128177.1(THRB):c.1012C>T (p.Arg338Trp)	THRB	3	24127631	Thyroid hormone resistance, generalized, autosomal dominant;Thyroid hormone resistance, selective pituitary
NM_001128177.1(THRB):c.947G>A (p.Arg316His)	THRB	3	24127696	Thyroid hormone resistance, selective pituitary
NM_000069.2(CACNA1S):c.-476G>A	CACNA1S	1	201112815	Thyrotoxic periodic paralysis
NM_000069.2(CACNA1S):c.258+57G>A	CACNA1S	1	201110107	Thyrotoxic periodic paralysis
NM_001194958.2(KCNJ18):c.127C>T (p.Arg43Cys)	KCNJ18	17	21702913	Thyrotoxic periodic paralysis
NM_001194958.2(KCNJ18):c.419C>T (p.Thr140Met)	KCNJ18	17	21703205	Thyrotoxic periodic paralysis

NM_001194958.2(KCNJ18):c.502G>A (p.Val168Met)	KCNJ18	17	21703288	Thyrotoxic periodic paralysis	
NM_001194958.2(KCNJ18):c.1195C>T (p.Arg399Ter)	KCNJ18	17	21703981	Thyrotoxic periodic paralysis	
NM_001194958.2(KCNJ18):c.1219C>T (p.Gln407Ter)	KCNJ18	17	21704005	Thyrotoxic periodic paralysis	
NM_001194958.2(KCNJ18):c.614G>A (p.Arg205His)	KCNJ18	17	21703400	Thyrotoxic periodic paralysis 2	
NM_001194958.2(KCNJ18):c.1061C>T (p.Thr354Met)	KCNJ18	17	21703847	Thyrotoxic periodic paralysis;Thyrotoxic periodic paralysis 2	
NM_003301.5(TRHR):c.49C>T (p.Arg17Ter)	TRHR	8	109087561	Thyrotropin-releasing hormone resistance, generalized	
NM_022458.3(LMBR1):c.423+4917G>A	LMBR1	7	156791472	Tibia, hypoplasia or aplasia of, with polydactyly	
NM_001167625.1(CACNA1C):c.1216G>A (p.Gly406Arg)	CACNA1C	12	2504538	Timothy syndrome;not provided	
NM_000594.3(TNF):c.322C>T (p.Arg108Trp)	TNF	6	31577157	TNF receptor binding, altered	
NM_001065.3(TNFRSF1A):c.185G>A (p.Cys62Tyr)	TNFRSF1A	12	6334099	TNF receptor-associated periodic fever syndrome (TRAPS)	
NM_001065.3(TNFRSF1A):c.350G>A (p.Cys117Tyr)	TNFRSF1A	12	6333489	TNF receptor-associated periodic fever syndrome (TRAPS)	
NM_001065.3(TNFRSF1A):c.236C>T (p.Thr79Met)	TNFRSF1A	12	6333823	TNF receptor-associated periodic fever syndrome (TRAPS);not provided	
NM_006194.3(PAX9):c.76C>T (p.Arg26Trp)	PAX9	14	36662968	Tooth agenesis, selective, 3	
NM_006194.3(PAX9):c.151G>A (p.Gly51Ser)	PAX9	14	36663043	Tooth agenesis, selective, 3	
NM_006194.3(PAX9):c.139C>T (p.Arg47Trp)	PAX9	14	36663031	Tooth agenesis, selective, 3	
NM_002336.2(LRP6):c.56C>T (p.Ala19Val)	LRP6	12	12244655	Tooth agenesis, selective, 7	
NM_003394.3(WNT10B):c.632G>A (p.Arg211Gln)	WNT10B	12	48968025	Tooth agenesis, selective, 8	
NM_003394.3(WNT10B):c.786G>A (p.Trp262Ter)	WNT10B	12	48966479	Tooth agenesis, selective, 8	
NM_022469.3(GREM2):c.38C>T (p.Ala13Val)	GREM2	1	240493438	TOOTH AGENESIS, SELECTIVE, 9	
NM_001399.4(EDA):c.1013C>T (p.Thr338Met)	EDA	X	70035446	Tooth agenesis, selective, X-linked, 1	
NM_001399.4(EDA):c.865C>T (p.Arg289Cys)	EDA	X	70033469	Tooth agenesis, selective, X-linked, 1	
NM_001399.4(EDA):c.1001G>A (p.Arg334His)	EDA	X	70035434	Tooth agenesis, selective, X-linked, 1	
NM_052910.2(SLITRK1):c.*689G>A	SLITRK1	13	83878728	Tourette Syndrome	
NM_002112.3(HDC):c.951G>A (p.Trp317Ter)	HDC	15	50252520	Tourette Syndrome	
NM_002968.2(SALL1):c.826C>T (p.Arg276Ter)	SALL1	16	51141396	Townes syndrome	
NM_002968.2(SALL1):c.949C>T (p.Pro317Ser)	SALL1	16	51141273	Townes syndrome	
NM_002968.2(SALL1):c.3160C>T (p.Arg1054Ter)	SALL1	16	51139062	Townes syndrome	
NM_002968.2(SALL1):c.727C>T (p.Gln243Ter)	SALL1	16	51141495	Townes syndrome	
NM_002968.2(SALL1):c.967C>T (p.Gln323Ter)	SALL1	16	51141255	Townes-Brocks-branchiootorenal-like syndrome	
NM_000094.3(COL7A1):c.4556G>A (p.Gly1519Asp)	COL7A1	3	48582616	Transient bullous dermolysis of the newborn	
NM_000094.3(COL7A1):c.4565G>A (p.Gly1522Glu)	COL7A1	3	48582512	Transient bullous dermolysis of the newborn	
NM_001109809.2(ZFP57):c.743G>A (p.Arg248His)	ZFP57	6	29673368	Transient neonatal diabetes mellitus 1	
NM_000352.4(ABCC8):c.3544C>T (p.Arg1182Trp)	ABCC8	11	17404525	Transient neonatal diabetes mellitus 2	
NM_000525.3(KCNJ11):c.157G>A (p.Gly53Ser)	KCNJ11	11	17387935	Transient neonatal diabetes mellitus 3	
NM_015335.4(MED13L):c.5615G>A (p.Arg1872His)	MED13L	12	115975287	Transposition of great arteries	
NM_000356.3(TCOF1):c.754C>T (p.Gln252Ter)	TCOF1	5	150374288	Treacher Collins syndrome 1	
NM_000356.3(TCOF1):c.2731C>T (p.Arg911Ter)	TCOF1	5	150388004	Treacher Collins syndrome 1	
NM_004960.3(FUS):c.1292C>T (p.Pro431Leu)	FUS	16	31190398	Tremor, hereditary essential, 4	
NM_004960.3(FUS):c.868C>T (p.Gln290Ter)	FUS	16	31189158	Tremor, hereditary essential, 4	
NM_001098816.2(TENM4):c.4324G>A (p.Ala1442Thr)	TENM4	11	78702289	Tremor, hereditary essential, 5	
NM_001098816.2(TENM4):c.3412G>A (p.Val1138Met)	TENM4	11	78726217	Tremor, hereditary essential, 5	
NM_014639.3(TTC37):c.439C>T (p.Gln147Ter)	TTC37	5	95540794	Trichohepatoenteric syndrome 1	
NM_014639.3(TTC37):c.2251C>T (p.Gln751Ter)	TTC37	5	95517186	Trichohepatoenteric syndrome 1	
NM_014639.3(TTC37):c.2808G>A (p.Trp936Ter)	TTC37	5	95512589	Trichohepatoenteric syndrome 1	
NM_006702.4(PNPLA6):c.3382G>A (p.Gly1128Ser)	PNPLA6	19	7558948	Trichomegaly with mental retardation, dwarfism and pigmentary degeneration of retina	

NM_006702.4(PNPLA6):c.3152G>A (p.Arg1051Gln)	PNPLA6	19	7556710	Trichomegaly with mental retardation, dwarfism and pigmentary degeneration of retina
NM_014112.4(TRPS1):c.2557C>T (p.Arg853Ter)	TRPS1	8	115587144	Trichorhinophalangeal dysplasia type I
NM_014112.4(TRPS1):c.2893C>T (p.Arg965Cys)	TRPS1	8	115415015	Trichorhinophalangeal dysplasia type I
NM_014112.4(TRPS1):c.2894G>A (p.Arg965His)	TRPS1	8	115415014	Trichorhinophalangeal dysplasia type I
NM_014112.4(TRPS1):c.1693C>T (p.Gln565Ter)	TRPS1	8	115604276	Trichorhinophalangeal dysplasia type I
NM_014112.4(TRPS1):c.1630C>T (p.Arg544Ter)	TRPS1	8	115604339	Trichorhinophalangeal dysplasia type I
NM_014112.4(TRPS1):c.1870C>T (p.Arg624Ter)	TRPS1	8	115604099	Trichorhinophalangeal dysplasia type I;not provided
NM_014112.4(TRPS1):c.2762G>A (p.Arg921Gln)	TRPS1	8	115418391	Trichorhinophalangeal syndrome type 3
NM_014112.4(TRPS1):c.2795C>T (p.Ala932Val)	TRPS1	8	115418358	Trichorhinophalangeal syndrome type 3
NM_000400.3(ERCC2):c.1972C>T (p.Arg658Cys)	ERCC2	19	45352580	Trichothiodystrophy 1, photosensitive
NM_000400.3(ERCC2):c.2164C>T (p.Arg722Trp)	ERCC2	19	45352235	Trichothiodystrophy 1, photosensitive;not provided
NM_207118.2(GTF2H5):c.166C>T (p.Arg56Ter)	GTF2H5	6	158192107	Trichothiodystrophy 1, photosensitive;Trichothiodystrophy 3, photosensitive
NM_000400.3(ERCC2):c.335G>A (p.Arg112His)	ERCC2	19	45368655	Trichothiodystrophy 1, photosensitive;Xeroderma pigmentosum, group D;not provided
NM_006978.2(RNF113A):c.901C>T (p.Gln301Ter)	RNF113A	X	119870713	Trichothiodystrophy 5, nonphotosensitive
NM_138701.3(MPLKIP):c.339+1G>A	MPLKIP	7	40134228	Trichothiodystrophy, nonphotosensitive 1
NM_016006.4(ABHD5):c.19G>A (p.Glu7Lys)	ABHD5	3	43691011	Triglyceride storage disease with ichthyosis
NM_016006.4(ABHD5):c.778G>A (p.Glu260Lys)	ABHD5	3	43717675	Triglyceride storage disease with ichthyosis
NM_001002294.2(FMO3):c.154G>A (p.Ala52Thr)	FMO3	1	171103806	Trimethylaminuria
NM_006894.5(FMO3):c.458C>T (p.Pro153Leu)	FMO3	1	171107811	Trimethylaminuria
NM_006894.5(FMO3):c.1474C>T (p.Arg492Trp)	FMO3	1	171117317	Trimethylaminuria
NM_001002294.2(FMO3):c.1302G>A (p.Met434Ile)	FMO3	1	171117145	Trimethylaminuria
NM_001002294.2(FMO3):c.94G>A (p.Glu32Lys)	FMO3	1	171092752	Trimethylaminuria
NM_001159287.1(TPI1):c.236G>A (p.Cys79Tyr)	TPI1	12	6868873	Triosephosphate isomerase deficiency
NM_001159287.1(TPI1):c.478G>A (p.Gly160Arg)	TPI1	12	6869300	Triosephosphate isomerase manchester
NM_001708.2(OPN1SW):c.235G>A (p.Gly79Arg)	OPN1SW	7	128775556	Tritanopia
NM_001708.2(OPN1SW):c.790C>T (p.Pro264Ser)	OPN1SW	7	128773786	Tritanopia
NM_000368.4(TSC1):c.2341C>T (p.Gln781Ter)	TSC1	9	132902655	Tuberous sclerosis 1;Tuberous sclerosis syndrome
NM_000368.4(TSC1):c.2041+1G>A	TSC1	9	132904410	Tuberous sclerosis 1;Tuberous sclerosis syndrome
NM_000368.4(TSC1):c.2698C>T (p.Gln900Ter)	TSC1	9	132897538	Tuberous sclerosis 1;Tuberous sclerosis syndrome
NM_000368.4(TSC1):c.1498C>T (p.Arg500Ter)	TSC1	9	132906080	Tuberous sclerosis 1;Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.682C>T (p.Arg228Ter)	TSC1	9	132921418	Tuberous sclerosis 1;Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.733C>T (p.Arg245Ter)	TSC1	9	132921367	Tuberous sclerosis 1;Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.973C>T (p.Gln325Ter)	TSC1	9	132911509	Tuberous sclerosis 1;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.1474C>T (p.Gln492Ter)	TSC2	16	2064302	Tuberous sclerosis 2
NM_000548.4(TSC2):c.1432C>T (p.Gln478Ter)	TSC2	16	2063042	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.1322G>A (p.Trp441Ter)	TSC2	16	2062561	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.4846C>T (p.Gln1616Ter)	TSC2	16	2086376	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.648+1G>A	TSC2	16	2056245	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.4989+1G>A	TSC2	16	2086872	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.4318C>T (p.Gln1440Ter)	TSC2	16	2084540	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.2666C>T (p.Ala889Val)	TSC2	16	2076094	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.1599+1G>A	TSC2	16	2064428	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.2108G>A (p.Trp703Ter)	TSC2	16	2072251	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.848+281C>T	TSC2	16	2057459	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.5161-1G>A	TSC2	16	2088226	Tuberous sclerosis 2;Tuberous sclerosis syndrome

NM_000548.4(TSC2):c.5170C>T (p.Gln1724Ter)	TSC2	16	2088236	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.336+1G>A	TSC2	16	2053453	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.4762C>T (p.Gln1588Ter)	TSC2	16	2086292	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.2251C>T (p.Arg751Ter)	TSC2	16	2072879	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.1255C>T (p.Pro419Ser)	TSC2	16	2062006	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.2688G>A (p.Trp896Ter)	TSC2	16	2076116	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.3623G>A (p.Trp1208Ter)	TSC2	16	2081607	Tuberous sclerosis 2;Tuberous sclerosis syndrome
NM_000548.4(TSC2):c.2713C>T (p.Arg905Trp)	TSC2	16	2076141	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.1372C>T (p.Arg458Ter)	TSC2	16	2062982	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.3094C>T (p.Arg1032Ter)	TSC2	16	2079159	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.3412C>T (p.Arg1138Ter)	TSC2	16	2080179	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.976-15G>A	TSC2	16	2060655	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.5160+1G>A	TSC2	16	2088140	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.5227C>T (p.Arg1743Trp)	TSC2	16	2088293	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.2098-1G>A	TSC2	16	2072240	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.3598C>T (p.Arg1200Trp)	TSC2	16	2080365	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.848+1G>A	TSC2	16	2057179	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.5228G>A (p.Arg1743Gln)	TSC2	16	2088294	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.4375C>T (p.Arg1459Ter)	TSC2	16	2084597	Tuberous sclerosis 2;Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.1963C>T (p.Gln655Ter)	TSC1	9	132905615	Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.1997+1G>A	TSC1	9	132905580	Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.2227C>T (p.Gln743Ter)	TSC1	9	132902769	Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.2689C>T (p.Gln897Ter)	TSC1	9	132897547	Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.737+1G>A	TSC1	9	132921362	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.3532C>T (p.Gln1178Ter)	TSC2	16	2080299	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.4573C>T (p.Gln1525Ter)	TSC2	16	2085233	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.4829G>A (p.Trp1610Ter)	TSC2	16	2086359	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.4993C>T (p.Gln1665Ter)	TSC2	16	2087866	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.5219G>A (p.Trp1740Ter)	TSC2	16	2088285	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.5254C>T (p.Gln1752Ter)	TSC2	16	2088320	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.849-1G>A	TSC2	16	2058746	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.1831C>T (p.Arg611Trp)	TSC2	16	2070570	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.268C>T (p.Gln90Ter)	TSC2	16	2053384	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.496C>T (p.Gln166Ter)	TSC2	16	2055416	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.774+1G>A	TSC2	16	2056770	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.3397+5G>A	TSC2	16	2079674	Tuberous sclerosis syndrome;not provided
NM_000548.4(TSC2):c.2194C>T (p.Gln732Ter)	TSC2	16	2072337	Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.2272C>T (p.Gln758Ter)	TSC1	9	132902724	Tuberous sclerosis syndrome;not provided
NM_000368.4(TSC1):c.2389C>T (p.Gln797Ter)	TSC1	9	132902607	Tuberous sclerosis syndrome;not provided
NM_004656.3(BAP1):c.799C>T (p.Gln267Ter)	BAP1	3	52405897	Tumor susceptibility linked to germline BAP1 mutations
NM_004656.3(BAP1):c.778C>T (p.Gln260Ter)	BAP1	3	52406258	Tumor susceptibility linked to germline BAP1 mutations
NM_004656.3(BAP1):c.2050C>T (p.Gln684Ter)	BAP1	3	52402608	Tumor susceptibility linked to germline BAP1 mutations;not provided
NM_020638.2(FGF23):c.386C>T (p.Ser129Phe)	FGF23	12	4370713	Tumoral calcinosis, familial, hyperphosphatemic
NM_004482.3(GALNT3):c.1524+1G>A	GALNT3	2	165754931	Tumoral calcinosis, familial, hyperphosphatemic
NM_004482.3(GALNT3):c.484C>T (p.Arg162Ter)	GALNT3	2	165770217	Tumoral calcinosis, familial, hyperphosphatemic

NM_004482.3(GALNT3):c.1524+5G>A	GALNT3	2	165754927	Tumoral calcinosis, familial, hyperphosphatemic
NM_004482.3(GALNT3):c.1774C>T (p.Gln592Ter)	GALNT3	2	165749747	Tumoral calcinosis, familial, hyperphosphatemic
NM_004482.3(GALNT3):c.1441C>T (p.Gln481Ter)	GALNT3	2	165755015	Tumoral calcinosis, familial, hyperphosphatemic
NM_004482.3(GALNT3):c.1525_1626del	GALNT3	2	165754626	Tumoral calcinosis, familial, hyperphosphatemic
NM_000372.4(TYR):c.272G>A (p.Cys91Tyr)	TYR	11	89178225	Tyrosinase-negative oculocutaneous albinism
NM_000372.4(TYR):c.658C>T (p.Gln220Ter)	TYR	11	89178611	Tyrosinase-negative oculocutaneous albinism
NM_000372.4(TYR):c.661G>A (p.Glu221Lys)	TYR	11	89178614	Tyrosinase-negative oculocutaneous albinism
NM_000372.4(TYR):c.982G>A (p.Glu328Lys)	TYR	11	89191364	Tyrosinase-negative oculocutaneous albinism
NM_000372.4(TYR):c.1064C>T (p.Ala355Val)	TYR	11	89227850	Tyrosinase-negative oculocutaneous albinism
NM_000372.4(TYR):c.230G>A (p.Arg77Gln)	TYR	11	89178183	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.533G>A (p.Trp178Ter)	TYR	11	89178486	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.164G>A (p.Cys55Tyr)	TYR	11	89178117	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.616G>A (p.Ala206Thr)	TYR	11	89178569	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.1255G>A (p.Gly419Arg)	TYR	11	89284843	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.61C>T (p.Pro21Ser)	TYR	11	89178014	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.649C>T (p.Arg217Trp)	TYR	11	89178602	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.896G>A (p.Arg299His)	TYR	11	89191278	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.1336G>A (p.Gly446Ser)	TYR	11	89284924	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.1342G>A (p.Asp448Asn)	TYR	11	89284930	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.707G>A (p.Trp236Ter)	TYR	11	89178660	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.1204C>T (p.Arg402Ter)	TYR	11	89284792	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.229C>T (p.Arg77Trp)	TYR	11	89178182	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000372.4(TYR):c.650G>A (p.Arg217Gln)	TYR	11	89178603	Tyrosinase-negative oculocutaneous albinism;not provided
NM_000275.2(OCA2):c.1001C>T (p.Ala334Val)	OCA2	15	28014819	Tyrosinase-positive oculocutaneous albinism
NM_000275.2(OCA2):c.1182G>A (p.Met394Ile)	OCA2	15	27989601	Tyrosinase-positive oculocutaneous albinism
NM_000275.2(OCA2):c.2228C>T (p.Pro743Leu)	OCA2	15	27871170	Tyrosinase-positive oculocutaneous albinism;not provided
NM_000275.2(OCA2):c.1503+5G>A	OCA2	15	27983340	Tyrosinase-positive oculocutaneous albinism;not provided
NM_003331.4(TYK2):c.1912C>T (p.Arg638Ter)	TYK2	19	10361817	Tyrosine kinase 2 deficiency
NM_000353.2(TAT):c.169C>T (p.Arg57Ter)	TAT	16	71576247	Tyrosinemia type 2
NM_000353.2(TAT):c.1249C>T (p.Arg417Ter)	TAT	16	71568260	Tyrosinemia type 2
NM_000137.2(FAH):c.1009G>A (p.Gly337Ser)	FAH	15	80180172	Tyrosinemia type I
NM_000137.2(FAH):c.786G>A (p.Trp262Ter)	FAH	15	80173093	Tyrosinemia type I
NM_000137.2(FAH):c.782C>T (p.Pro261Leu)	FAH	15	80173089	Tyrosinemia type I;not provided
NM_000137.2(FAH):c.456G>A (p.Trp152Ter)	FAH	15	80168052	Tyrosinemia type I;not provided
NM_000137.2(FAH):c.520C>T (p.Arg174Ter)	FAH	15	80168116	Tyrosinemia type I;not provided
NM_000403.3(GALE):c.269G>A (p.Gly90Glu)	GALE	1	23798199	UDPglucose-4-epimerase deficiency
NM_000403.3(GALE):c.505C>T (p.Arg169Trp)	GALE	1	23797718	UDPglucose-4-epimerase deficiency
NM_000403.3(GALE):c.715C>T (p.Arg239Trp)	GALE	1	23796777	UDPglucose-4-epimerase deficiency
NM_000403.3(GALE):c.905G>A (p.Gly302Asp)	GALE	1	23796234	UDPglucose-4-epimerase deficiency
NM_000403.3(GALE):c.956G>A (p.Gly319Glu)	GALE	1	23796183	UDPglucose-4-epimerase deficiency;not provided
NM_004369.3(COL6A3):c.1393C>T (p.Arg465Ter)	COL6A3	2	237381419	Ullrich congenital muscular dystrophy 1
NM_001849.3(COL6A2):c.1493G>A (p.Arg498His)	COL6A2	21	46121590	Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.841G>A (p.Gly281Arg)	COL6A1	21	45989120	Ullrich congenital muscular dystrophy 1
NM_001849.3(COL6A2):c.1870G>A (p.Glu624Lys)	COL6A2	21	46125518	Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.904G>A (p.Gly302Arg)	COL6A1	21	45989752	Ullrich congenital muscular dystrophy 1

NM_004369.3(COL6A3):c.6220G>A (p.Gly2074Ser)	COL6A3	2	237360150	Ullrich congenital muscular dystrophy 1
NM_001848.2(COL6A1):c.957+1G>A	COL6A1	21	45990285	Ullrich congenital muscular dystrophy 1
NM_004369.3(COL6A3):c.7024C>T (p.Arg2342Ter)	COL6A3	2	237347812	Ullrich congenital muscular dystrophy 1;not provided
NM_004370.5(COL12A1):c.7840+1G>A	COL12A1	6	75113601	Ullrich congenital muscular dystrophy 2
NM_004625.3(WNT7A):c.874C>T (p.Arg292Cys)	WNT7A	3	13819120	Ulna and fibula absence of with severe limb deficiency
NM_004625.3(WNT7A):c.610G>A (p.Gly204Ser)	WNT7A	3	13819384	Ulna and fibula absence of with severe limb deficiency
NM_004625.3(WNT7A):c.664C>T (p.Arg222Trp)	WNT7A	3	13819330	Ulna and fibula absence of with severe limb deficiency
NM_004625.3(WNT7A):c.214G>A (p.Glu72Lys)	WNT7A	3	13875031	Ulna and fibula absence of with severe limb deficiency
NM_004625.3(WNT7A):c.304C>T (p.Arg102Trp)	WNT7A	3	13854798	Ulna and fibula absence of with severe limb deficiency
NM_016569.3(TBX3):c.1051C>T (p.Gln351Ter)	TBX3	12	114676361	Ulnar-mammary syndrome
NM_016569.3(TBX3):c.1483C>T (p.Gln495Ter)	TBX3	12	114674452	Ulnar-mammary syndrome
NM_016233.2(PADI3):c.881C>T (p.Ala294Val)	PADI3	1	17270928	Uncombable hair syndrome
NM_007113.3(TCHH):c.991C>T (p.Gln331Ter)	TCHH	1	152112226	UNCOMBABLE HAIR SYNDROME 3
NM_000100.3(CSTB):c.202C>T (p.Arg68Ter)	CSTB	21	43774297	Unverricht-Lundborg syndrome
NM_000100.3(CSTB):c.168G>A (p.Lys56=)	CSTB	21	43774658	Unverricht-Lundborg syndrome
NM_000100.3(CSTB):c.66G>A (p.Gln22=)	CSTB	21	43776204	Unverricht-Lundborg syndrome
NM_000100.3(CSTB):c.149G>A (p.Gly50Glu)	CSTB	21	43774677	Unverricht-Lundborg syndrome
NM_000100.3(CSTB):c.136C>T (p.Gln46Ter)	CSTB	21	43774690	Unverricht-Lundborg syndrome;not provided
NM_139025.4(ADAMTS13):c.304C>T (p.Arg102Cys)	ADAMTS13	9	133424452	Upshaw-Schulman syndrome
NM_139025.4(ADAMTS13):c.587C>T (p.Thr196Ile)	ADAMTS13	9	133426246	Upshaw-Schulman syndrome
NM_139025.4(ADAMTS13):c.1193G>A (p.Arg398His)	ADAMTS13	9	133433478	Upshaw-Schulman syndrome
NM_139025.4(ADAMTS13):c.3638G>A (p.Cys1213Tyr)	ADAMTS13	9	133456138	Upshaw-Schulman syndrome
NM_139025.4(ADAMTS13):c.2074C>T (p.Arg692Cys)	ADAMTS13	9	133442504	Upshaw-Schulman syndrome
NM_139025.4(ADAMTS13):c.1345C>T (p.Gln449Ter)	ADAMTS13	9	133436865	Upshaw-Schulman syndrome
NM_139025.4(ADAMTS13):c.414+1G>A	ADAMTS13	9	133425613	Upshaw-Schulman syndrome
NM_139025.4(ADAMTS13):c.749C>T (p.Ala250Val)	ADAMTS13	9	133428696	Upshaw-Schulman syndrome
NM_139025.4(ADAMTS13):c.331-1G>A	ADAMTS13	9	133425528	Upshaw-Schulman syndrome
NM_001002010.2(NT5C3A):c.646C>T (p.Gln216Ter)	NT5C3A	7	33017501	Uridine 5-prime monophosphate hydrolase deficiency, hemolytic anemia due to
NM_001165974.1(UROC1):c.1528C>T (p.Arg510Cys)	UROC1	3	126498141	Urocanate hydratase deficiency
NM_014813.2(LRIG2):c.2125C>T (p.Arg709Ter)	LRIG2	1	113114471	Urofacial syndrome 2
NM_000260.3(MYO7A):c.1200+1G>A	MYO7A	11	77160283	Usher syndrome, type 1
NM_000260.3(MYO7A):c.141G>A (p.Trp47Ter)	MYO7A	11	77147806	Usher syndrome, type 1
NM_000260.3(MYO7A):c.1556G>A (p.Gly519Asp)	MYO7A	11	77162854	Usher syndrome, type 1
NM_000260.3(MYO7A):c.1900C>T (p.Arg634Ter)	MYO7A	11	77172850	Usher syndrome, type 1
NM_000260.3(MYO7A):c.1963C>T (p.Gln655Ter)	MYO7A	11	77174783	Usher syndrome, type 1
NM_000260.3(MYO7A):c.2094+1G>A	MYO7A	11	77174915	Usher syndrome, type 1
NM_000260.3(MYO7A):c.3508G>A (p.Glu1170Lys)	MYO7A	11	77189348	Usher syndrome, type 1
NM_000260.3(MYO7A):c.4293G>A (p.Trp1431Ter)	MYO7A	11	77194494	Usher syndrome, type 1
NM_000260.3(MYO7A):c.5101C>T (p.Arg1701Ter)	MYO7A	11	77202357	Usher syndrome, type 1
NM_000260.3(MYO7A):c.5617C>T (p.Arg1873Trp)	MYO7A	11	77205598	Usher syndrome, type 1
NM_000260.3(MYO7A):c.5660C>T (p.Pro1887Leu)	MYO7A	11	77206120	Usher syndrome, type 1
NM_000260.3(MYO7A):c.6070C>T (p.Arg2024Ter)	MYO7A	11	77211170	Usher syndrome, type 1
NM_000260.3(MYO7A):c.640G>A (p.Gly214Arg)	MYO7A	11	77156909	Usher syndrome, type 1
NM_000260.3(MYO7A):c.470+1G>A	MYO7A	11	77156092	Usher syndrome, type 1
NM_000260.3(MYO7A):c.5968C>T (p.Gln1990Ter)	MYO7A	11	77208720	Usher syndrome, type 1

NM_033056.3(PCDH15):c.158-1G>A	PCDH15	10	54378943	Usher syndrome, type 1
NM_000260.3(MYO7A):c.3719G>A (p.Arg1240Gln)	MYO7A	11	77190108	Usher syndrome, type 1;not provided
NM_000260.3(MYO7A):c.494C>T (p.Thr165Met)	MYO7A	11	77156683	Usher syndrome, type 1;not provided
NM_000260.3(MYO7A):c.5392C>T (p.Gln1798Ter)	MYO7A	11	77204141	Usher syndrome, type 1;not provided
NM_000260.3(MYO7A):c.5648G>A (p.Arg1883Gln)	MYO7A	11	77206108	Usher syndrome, type 1;not provided
NM_000260.3(MYO7A):c.448C>T (p.Arg150Ter)	MYO7A	11	77156069	Usher syndrome, type 1;Usher syndrome, type 1B
NM_000260.3(MYO7A):c.700C>T (p.Gln234Ter)	MYO7A	11	77156969	Usher syndrome, type 1;Usher syndrome, type 1B
NM_000260.3(MYO7A):c.635G>A (p.Arg212His)	MYO7A	11	77156904	Usher syndrome, type 1;Usher syndrome, type 1B
NM_000260.3(MYO7A):c.1996C>T (p.Arg666Ter)	MYO7A	11	77174816	Usher syndrome, type 1;Usher syndrome, type 1B
NM_005709.3(USH1C):c.216G>A (p.Val72=)	USH1C	11	17531431	Usher syndrome, type 1;Usher syndrome, type 1C
NM_000260.3(MYO7A):c.634C>T (p.Arg212Cys)	MYO7A	11	77156903	Usher syndrome, type 1B
NM_005709.3(USH1C):c.91C>T (p.Arg31Ter)	USH1C	11	17533268	Usher syndrome, type 1C
NM_005709.3(USH1C):c.308G>A (p.Arg103His)	USH1C	11	17531233	Usher syndrome, type 1C
NM_005709.3(USH1C):c.7C>T (p.Arg3Ter)	USH1C	11	17544301	Usher syndrome, type 1C
NM_022124.5(CDH23):c.7362+5G>A	CDH23	10	71799634	Usher syndrome, type 1D
NM_022124.5(CDH23):c.3880C>T (p.Gln1294Ter)	CDH23	10	71732151	Usher syndrome, type 1D
NM_022124.5(CDH23):c.3481C>T (p.Arg1161Ter)	CDH23	10	71725422	Usher syndrome, type 1D
NM_022124.5(CDH23):c.3628C>T (p.Gln1210Ter)	CDH23	10	71730517	Usher syndrome, type 1D
NM_022124.5(CDH23):c.5272C>T (p.Gln1758Ter)	CDH23	10	71779351	Usher syndrome, type 1D
NM_022124.5(CDH23):c.5712+1G>A	CDH23	10	71785101	Usher syndrome, type 1D
NM_022124.5(CDH23):c.5712G>A (p.Thr1904=)	CDH23	10	71785100	Usher syndrome, type 1D
NM_022124.5(CDH23):c.5923+1G>A	CDH23	10	71789043	Usher syndrome, type 1D
NM_022124.5(CDH23):c.6049+1G>A	CDH23	10	71790414	Usher syndrome, type 1D
NM_022124.5(CDH23):c.7776G>A (p.Trp2592Ter)	CDH23	10	71803324	Usher syndrome, type 1D
NM_001142763.1(PCDH15):c.2986C>T (p.Arg996Ter)	PCDH15	10	53961790	Usher syndrome, type 1D
NM_022124.5(CDH23):c.9556C>T (p.Arg3186Ter)	CDH23	10	71812813	Usher syndrome, type 1D
NM_022124.5(CDH23):c.1987-1G>A	CDH23	10	71687646	Usher syndrome, type 1D
NM_022124.5(CDH23):c.3706C>T (p.Arg1236Ter)	CDH23	10	71730595	Usher syndrome, type 1D;not provided
NM_022124.5(CDH23):c.4309C>T (p.Arg1437Ter)	CDH23	10	71738597	Usher syndrome, type 1D;not provided
NM_022124.5(CDH23):c.6050-9G>A	CDH23	10	71791123	Usher syndrome, type 1D;not provided
NM_033056.3(PCDH15):c.7C>T (p.Arg3Ter)	PCDH15	10	54664256	Usher syndrome, type 1F
NM_033056.3(PCDH15):c.3316C>T (p.Arg1106Ter)	PCDH15	10	53938872	Usher syndrome, type 1F
NM_033056.3(PCDH15):c.1927C>T (p.Arg643Ter)	PCDH15	10	54090054	Usher syndrome, type 1F
NM_001142772.1(PCDH15):c.400C>T (p.Arg134Ter)	PCDH15	10	54369194	Usher syndrome, type 1F
NM_033056.3(PCDH15):c.3358C>T (p.Arg1120Ter)	PCDH15	10	53938830	Usher syndrome, type 1F
NM_173477.4(USH1G):c.113G>A (p.Trp38Ter)	USH1G	17	74922961	Usher syndrome, type 1G
NM_206933.2(USH2A):c.11048-1G>A	USH2A	1	215759844	Usher syndrome, type 2A
NM_206933.2(USH2A):c.1143+1G>A	USH2A	1	216325304	Usher syndrome, type 2A
NM_206933.2(USH2A):c.11954G>A (p.Trp3985Ter)	USH2A	1	215728142	Usher syndrome, type 2A
NM_206933.2(USH2A):c.12868C>T (p.Gln4290Ter)	USH2A	1	215675043	Usher syndrome, type 2A
NM_206933.2(USH2A):c.14180G>A (p.Trp4727Ter)	USH2A	1	215650755	Usher syndrome, type 2A
NM_206933.2(USH2A):c.14911C>T (p.Arg4971Ter)	USH2A	1	215640615	Usher syndrome, type 2A
NM_206933.2(USH2A):c.5788C>T (p.Arg1930Ter)	USH2A	1	216072958	Usher syndrome, type 2A
NM_206933.2(USH2A):c.5858-1G>A	USH2A	1	216070293	Usher syndrome, type 2A
NM_206933.2(USH2A):c.6224G>A (p.Trp2075Ter)	USH2A	1	216046532	Usher syndrome, type 2A

NM_206933.2(USH2A):c.820C>T (p.Arg274Ter)	USH2A	1	216327619	Usher syndrome, type 2A
NM_206933.2(USH2A):c.8981G>A (p.Trp2994Ter)	USH2A	1	215845898	Usher syndrome, type 2A
NM_206933.2(USH2A):c.9304C>T (p.Gln3102Ter)	USH2A	1	215838058	Usher syndrome, type 2A
NM_206933.2(USH2A):c.13010C>T (p.Thr4337Met)	USH2A	1	215674901	Usher syndrome, type 2A
NM_206933.2(USH2A):c.2983C>T (p.Gln995Ter)	USH2A	1	216231963	Usher syndrome, type 2A
NM_206933.2(USH2A):c.14248C>T (p.Gln4750Ter)	USH2A	1	215650687	Usher syndrome, type 2A
NM_206933.2(USH2A):c.6398G>A (p.Trp2133Ter)	USH2A	1	216000490	Usher syndrome, type 2A
NM_206933.2(USH2A):c.632G>A (p.Trp211Ter)	USH2A	1	216418533	Usher syndrome, type 2A
NM_206933.2(USH2A):c.6601C>T (p.Gln2201Ter)	USH2A	1	215998943	Usher syndrome, type 2A
NM_206933.2(USH2A):c.13316C>T (p.Thr4439Ile)	USH2A	1	215674595	Usher syndrome, type 2A
NM_206933.2(USH2A):c.4405C>T (p.Gln1469Ter)	USH2A	1	216175474	Usher syndrome, type 2A
NM_206933.2(USH2A):c.9570+1G>A	USH2A	1	215816996	Usher syndrome, type 2A
NM_206933.2(USH2A):c.8740C>T (p.Arg2914Ter)	USH2A	1	215867112	Usher syndrome, type 2A
NM_206933.2(USH2A):c.8681+1G>A	USH2A	1	215877757	Usher syndrome, type 2A
NM_206933.2(USH2A):c.1000C>T (p.Arg334Trp)	USH2A	1	216325448	Usher syndrome, type 2A
NM_206933.2(USH2A):c.14175G>A (p.Trp4725Ter)	USH2A	1	215650760	Usher syndrome, type 2A
NM_206933.2(USH2A):c.9390G>A (p.Trp3130Ter)	USH2A	1	215817177	Usher syndrome, type 2A
NM_206933.2(USH2A):c.908G>A (p.Arg303His)	USH2A	1	216325540	Usher syndrome, type 2A
NM_206933.2(USH2A):c.11156G>A (p.Arg3719His)	USH2A	1	215759735	Usher syndrome, type 2A;not provided
NM_206933.2(USH2A):c.5776+1G>A	USH2A	1	216073096	Usher syndrome, type 2A;not provided
NM_032119.3(ADGRV1):c.2398C>T (p.Arg800Ter)	ADGRV1	5	90642886	Usher syndrome, type 2C
NM_032119.3(ADGRV1):c.7406G>A (p.Trp2469Ter)	ADGRV1	5	90694162	Usher syndrome, type 2C
NM_032119.3(ADGRV1):c.12631C>T (p.Arg4211Ter)	ADGRV1	5	90778008	Usher syndrome, type 2C
NM_032119.3(ADGRV1):c.7129C>T (p.Arg2377Ter)	ADGRV1	5	90692782	Usher syndrome, type 2C
NM_032119.3(ADGRV1):c.14885G>A (p.Trp4962Ter)	ADGRV1	5	90807650	Usher syndrome, type 2C
NM_032119.3(ADGRV1):c.2898G>A (p.Glu966=)	ADGRV1	5	90644869	Usher syndrome, type 2C
NM_032119.3(ADGRV1):c.10426G>A (p.Gly3476Arg)	ADGRV1	5	90728933	Usher syndrome, type 2C
NM_015404.3(WHRN):c.307C>T (p.Gln103Ter)	WHRN	9	114504495	Usher syndrome, type 2D
NM_015404.3(WHRN):c.1267C>T (p.Arg423Ter)	WHRN	9	114424483	Usher syndrome, type 2D
NM_174878.2(CLRN1):c.619C>T (p.Arg207Ter)	CLRN1	3	150928016	Usher syndrome, type 3A
NM_002386.3(MC1R):c.470C>T (p.Thr157Ile)	MC1R	16	89919728	Uv-induced skin damage, susceptibility to
NM_000124.3(ERCC6):c.229C>T (p.Arg77Ter)	ERCC6	10	49532736	UV-sensitive syndrome
NM_006147.3(IRF6):c.5C>T (p.Ala2Val)	IRF6	1	209801409	Van der Woude syndrome
NM_006147.3(IRF6):c.1137G>A (p.Trp379Ter)	IRF6	1	209789709	Van der Woude syndrome
NM_006147.3(IRF6):c.1198C>T (p.Arg400Trp)	IRF6	1	209788626	Van der Woude syndrome
NM_006147.3(IRF6):c.134G>A (p.Arg45Gln)	IRF6	1	209801280	Van der Woude syndrome
NM_006147.3(IRF6):c.1186C>T (p.Pro396Ser)	IRF6	1	209788638	Van der Woude syndrome
NM_006147.3(IRF6):c.145C>T (p.Gln49Ter)	IRF6	1	209801269	Van der Woude syndrome
NM_006147.3(IRF6):c.1199G>A (p.Arg400Gln)	IRF6	1	209788625	Van der Woude syndrome
NM_198173.2(GRHL3):c.893G>A (p.Arg298His)	GRHL3	1	24338044	Van der Woude syndrome 2
NM_198173.2(GRHL3):c.1171C>T (p.Arg391Cys)	GRHL3	1	24342238	Van der Woude syndrome 2
NM_006147.3(IRF6):c.1210G>A (p.Glu404Lys)	IRF6	1	209788614	Van der Woude syndrome;not provided
NM_024582.4(FAT4):c.11455C>T (p.Arg3819Ter)	FAT4	4	125452471	Van Maldergem syndrome 2
NM_024582.4(FAT4):c.13193G>A (p.Cys4398Tyr)	FAT4	4	125490015	Van Maldergem syndrome 2
NM_001122764.2(PPOX):c.502C>T (p.Arg168Cys)	PPOX	1	161168462	Variegate porphyria

NM_001122764.2(PPOX):c.175C>T (p.Arg59Trp)	PPOX	1	161167187	Variegate porphyria	
NM_001122764.2(PPOX):c.503G>A (p.Arg168His)	PPOX	1	161168463	Variegate porphyria	
NM_133171.4(ELMO2):c.1065+1G>A	ELMO2	20	46375235	Vascular malformation, primary intraosseous	
NM_001039350.2(DPP6):c.52-141059C>T	DPP6	7	154305155	Ventricular fibrillation, paroxysmal familial, 2	
NM_002052.4(GATA4):c.1075G>A (p.Glu359Lys)	GATA4	8	11757012	Ventricular septal defect 1	
NM_002052.4(GATA4):c.1325C>T (p.Ala442Val)	GATA4	8	11758471	Ventricular septal defect 1	
NM_002052.4(GATA4):c.127C>T (p.Arg43Trp)	GATA4	8	11708439	Ventricular septal defect 1	
NM_001035.2(RYR2):c.6982C>T (p.Pro2328Ser)	RYR2	1	237639068	Ventricular tachycardia, catecholaminergic polymorphic, 1	
NM_001035.2(RYR2):c.13489C>T (p.Arg4497Cys)	RYR2	1	237791441	Ventricular tachycardia, catecholaminergic polymorphic, 1;not provided	
NM_001035.2(RYR2):c.527G>A (p.Arg176Gln)	RYR2	1	237377386	Ventricular tachycardia, catecholaminergic polymorphic, 1;not provided	
NM_001035.2(RYR2):c.14876G>A (p.Arg4959Gln)	RYR2	1	237832619	Ventricular tachycardia, catecholaminergic polymorphic, 1;not provided	
NM_001232.3(CASQ2):c.97C>T (p.Arg33Ter)	CASQ2	1	115768445	Ventricular tachycardia, catecholaminergic polymorphic, 2;not provided	
NM_001010874.4(TECRL):c.331+1G>A	TECRL	4	64328511	Ventricular tachycardia, catecholaminergic polymorphic, 3	
NM_001010874.4(TECRL):c.587G>A (p.Arg196Gln)	TECRL	4	64309896	Ventricular tachycardia, catecholaminergic polymorphic, 3	
NM_006073.3(TRDN):c.613C>T (p.Gln205Ter)	TRDN	6	123503899	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness;not provided	
NM_173689.6(CRB2):c.2277G>A (p.Trp759Ter)	CRB2	9	123371419	Ventriculomegaly with cystic kidney disease	
NM_173689.6(CRB2):c.1897C>T (p.Arg633Trp)	CRB2	9	123370950	Ventriculomegaly with cystic kidney disease	
NM_078480.2(PUF60):c.505C>T (p.His169Tyr)	PUF60	8	143818378	Verheij syndrome	
NM_001130144.2(LTBP3):c.421C>T (p.Gln141Ter)	LTBP3	11	65554291	Verloes Bourguignon syndrome	
NM_000018.3(ACADVL):c.1246G>A (p.Ala416Thr)	ACADVL	17	7223707	Very long chain acyl-CoA dehydrogenase deficiency	
NM_000018.3(ACADVL):c.1349G>A (p.Arg450His)	ACADVL	17	7223984	Very long chain acyl-CoA dehydrogenase deficiency	
NM_000018.3(ACADVL):c.1226C>T (p.Thr409Met)	ACADVL	17	7223687	Very long chain acyl-CoA dehydrogenase deficiency	
NM_000018.3(ACADVL):c.1405C>T (p.Arg469Trp)	ACADVL	17	7224040	Very long chain acyl-CoA dehydrogenase deficiency	
NM_000018.3(ACADVL):c.1532+1G>A	ACADVL	17	7224244	Very long chain acyl-CoA dehydrogenase deficiency	
NM_000018.3(ACADVL):c.1357C>T (p.Arg453Ter)	ACADVL	17	7223992	Very long chain acyl-CoA dehydrogenase deficiency	
NM_000018.3(ACADVL):c.637G>A (p.Ala213Thr)	ACADVL	17	7221966	Very long chain acyl-CoA dehydrogenase deficiency	
NM_000018.3(ACADVL):c.1078_1182del105	ACADVL	17	7223238	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_000018.3(ACADVL):c.1837C>T (p.Arg613Trp)	ACADVL	17	7224966	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_000018.3(ACADVL):c.1322G>A (p.Gly441Asp)	ACADVL	17	7223865	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_000018.3(ACADVL):c.1679-6G>A	ACADVL	17	7224636	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_000018.3(ACADVL):c.779C>T (p.Thr260Met)	ACADVL	17	7222203	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_000018.3(ACADVL):c.1406G>A (p.Arg469Gln)	ACADVL	17	7224041	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_000018.3(ACADVL):c.520G>A (p.Val174Met)	ACADVL	17	7221580	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_000018.3(ACADVL):c.553G>A (p.Gly185Ser)	ACADVL	17	7221613	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_000018.3(ACADVL):c.1097G>A (p.Arg366His)	ACADVL	17	7223152	Very long chain acyl-CoA dehydrogenase deficiency;not provided	
NM_002942.4(ROBO2):c.3706G>A (p.Ala1236Thr)	ROBO2	3	77622378	Vesicoureteral reflux 2	
NM_019105.6(TNXB):c.9764C>T (p.Thr3255Ile)	TNXB	6	32048638	Vesicoureteral reflux 8	
NM_019105.6(TNXB):c.3991G>A (p.Gly1331Arg)	TNXB	6	32081419	Vesicoureteral reflux 8	
NM_013447.3(ADGRE2):c.1475G>A (p.Cys492Tyr)	ADGRE2	19	14755069	Vibratory urticaria	
NM_018055.4(NODAL):c.778G>A (p.Gly260Arg)	NODAL	10	70435399	Visceral heterotaxy 5, autosomal	
NM_001615.3(ACTG2):c.118C>T (p.Arg40Cys)	ACTG2	2	73901429	Visceral myopathy	
NM_001615.3(ACTG2):c.119G>A (p.Arg40His)	ACTG2	2	73901430	Visceral myopathy	
NM_001615.3(ACTG2):c.593G>A (p.Gly198Asp)	ACTG2	2	73913626	Visceral myopathy	
NM_001615.3(ACTG2):c.613G>A (p.Ala205Thr)	ACTG2	2	73913646	Visceral myopathy	

NM_001615.3(ACTG2):c.532C>T (p.Arg178Cys)	ACTG2	2	73913565	Visceral myopathy;not provided
NM_000785.3(CYP27B1):c.374G>A (p.Gly125Glu)	CYP27B1	12	57766019	Vitamin D-dependent rickets, type 1
NM_000785.3(CYP27B1):c.1144C>T (p.Pro382Ser)	CYP27B1	12	57764169	Vitamin D-dependent rickets, type 1
NM_000785.3(CYP27B1):c.589+1G>A	CYP27B1	12	57765296	Vitamin D-dependent rickets, type 1
NM_000785.3(CYP27B1):c.1226C>T (p.Thr409Ile)	CYP27B1	12	57763798	Vitamin D-dependent rickets, type 1
NM_000785.3(CYP27B1):c.1027C>T (p.Leu343Phe)	CYP27B1	12	57764487	Vitamin D-dependent rickets, type 1
NM_001017535.1(VDR):c.98G>A (p.Gly33Asp)	VDR	12	47879016	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.218G>A (p.Arg73Gln)	VDR	12	47865106	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.985G>A (p.Glu329Lys)	VDR	12	47846374	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.239G>A (p.Arg80Gln)	VDR	12	47865085	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.149G>A (p.Arg50Gln)	VDR	12	47865175	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.454C>T (p.Gln152Ter)	VDR	12	47857512	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.137G>A (p.Gly46Asp)	VDR	12	47878977	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.1171C>T (p.Arg391Cys)	VDR	12	47844859	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.88C>T (p.Arg30Ter)	VDR	12	47879026	Vitamin D-dependent rickets, type 2
NM_001017535.1(VDR):c.1036G>A (p.Val346Met)	VDR	12	47844994	Vitamin D-dependent rickets, type 2
NM_000785.3(CYP27B1):c.320G>A (p.Arg107His)	CYP27B1	12	57766073	Vitamin D-Dependent Rickets;Vitamin D-dependent rickets, type 1
NM_000785.3(CYP27B1):c.1166G>A (p.Arg389His)	CYP27B1	12	57764147	Vitamin D-Dependent Rickets;Vitamin D-dependent rickets, type 1;not provided
NM_024006.5(VKORC1):c.292C>T (p.Arg98Trp)	VKORC1	16	31091334	Vitamin k-dependent clotting factors, combined deficiency of, 2
NM_004183.3(BEST1):c.896G>A (p.Gly299Glu)	BEST1	11	61959526	Vitelliform macular dystrophy type 2;not provided
NM_004183.3(BEST1):c.25G>A (p.Val9Met)	BEST1	11	61951831	Vitelliform macular dystrophy type 2;not provided
NM_004183.3(BEST1):c.728C>T (p.Ala243Val)	BEST1	11	61958159	Vitelliform macular dystrophy type 2;not provided
NM_004183.3(BEST1):c.140G>A (p.Arg47His)	BEST1	11	61951946	Vitelliform macular dystrophy type 2;not provided
NM_004183.3(BEST1):c.652C>T (p.Arg218Cys)	BEST1	11	61957402	Vitelliform macular dystrophy type 2;not provided
NM_004183.3(BEST1):c.256G>A (p.Val86Met)	BEST1	11	61955726	Vitreoretinochoroidopathy
NM_001844.4(COL2A1):c.3914G>A (p.Gly1305Asp)	COL2A1	12	47974835	Vitreoretinopathy with phalangeal epiphyseal dysplasia
NM_000551.3(VHL):c.208G>A (p.Glu70Lys)	VHL	3	10142055	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.464-1G>A	VHL	3	10149786	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.194C>T (p.Ser65Leu)	VHL	3	10142041	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.257C>T (p.Pro86Leu)	VHL	3	10142104	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.217C>T (p.Gln73Ter)	VHL	3	10142064	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.332G>A (p.Ser111Asn)	VHL	3	10142179	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.340+1G>A	VHL	3	10142188	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.445G>A (p.Ala149Thr)	VHL	3	10146618	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.470C>T (p.Thr157Ile)	VHL	3	10149793	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.485G>A (p.Cys162Tyr)	VHL	3	10149808	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.394C>T (p.Gln132Ter)	VHL	3	10146567	Von Hippel-Lindau syndrome
NM_000551.3(VHL):c.256C>T (p.Pro86Ser)	VHL	3	10142103	Von Hippel-Lindau syndrome;not provided
NM_000551.3(VHL):c.263G>A (p.Trp88Ter)	VHL	3	10142110	Von Hippel-Lindau syndrome;not provided
NM_000551.3(VHL):c.490C>T (p.Gln164Ter)	VHL	3	10149813	Von Hippel-Lindau syndrome;not provided
NM_181457.3(PAX3):c.668G>A (p.Arg223Gln)	PAX3	2	222232202	Waardenburg syndrome
NM_000248.3(MITF):c.722G>A (p.Trp241Ter)	MITF	3	69959284	Waardenburg syndrome
NM_000248.3(MITF):c.808C>T (p.Arg270Ter)	MITF	3	69959370	Waardenburg syndrome
NM_000115.4(EDNRB):c.618G>A (p.Trp206Ter)	EDNRB	13	77903339	Waardenburg syndrome
NM_181457.3(PAX3):c.149C>T (p.Pro50Leu)	PAX3	2	222297150	Waardenburg syndrome type 1

NM_006941.3(SOX10):c.1129C>T (p.Gln377Ter)	SOX10	22	37973767	Waardenburg syndrome type 2E, with neurologic involvement;Waardenburg syndrome type 4C
NM_000115.4(EDNRB):c.757C>T (p.Arg253Ter)	EDNRB	13	77903200	Waardenburg syndrome type 4A
NM_006941.3(SOX10):c.470C>T (p.Ala157Val)	SOX10	22	37978094	Waardenburg syndrome type 4C
NM_004385.4(VCAN):c.4004-1G>A	VCAN	5	83537006	Wagner syndrome
NM_004385.4(VCAN):c.9265+1G>A	VCAN	5	83542269	Wagner syndrome
NM_001079802.1(FKTN):c.607C>T (p.Arg203Ter)	FKTN	9	105604452	Walker-Warburg congenital muscular dystrophy;not provided
NM_001172435.1(RAB3GAP1):c.748+1G>A	RAB3GAP1	2	135120919	Warburg micro syndrome 1
NM_001172435.1(RAB3GAP1):c.2011C>T (p.Arg671Ter)	RAB3GAP1	2	135150456	Warburg micro syndrome 1
NM_001172435.1(RAB3GAP1):c.1734G>A (p.Trp578Ter)	RAB3GAP1	2	135135743	Warburg micro syndrome 1
NM_001172435.1(RAB3GAP1):c.899+1G>A	RAB3GAP1	2	135126250	Warburg micro syndrome 1
NM_012414.3(RAB3GAP2):c.1434G>A (p.Trp478Ter)	RAB3GAP2	1	220191121	Warburg micro syndrome 2
NM_012414.3(RAB3GAP2):c.3637C>T (p.Arg1213Ter)	RAB3GAP2	1	220153976	Warburg micro syndrome 2
NM_144628.3(TBC1D20):c.199C>T (p.Arg67Ter)	TBC1D20	20	447946	Warburg micro syndrome 4
NM_144628.3(TBC1D20):c.292C>T (p.Gln98Ter)	TBC1D20	20	445095	Warburg micro syndrome 4
NM_144628.3(TBC1D20):c.672G>A (p.Trp224Ter)	TBC1D20	20	440344	Warburg micro syndrome 4
NM_030653.3(DDX11):c.788G>A (p.Arg263Gln)	DDX11	12	31089147	Warsaw breakage syndrome
NM_004456.4(EZH2):c.2080C>T (p.His694Tyr)	EZH2	7	148809340	Weaver syndrome
NM_004456.4(EZH2):c.394C>T (p.Pro132Ser)	EZH2	7	148829818	Weaver syndrome
NM_004456.4(EZH2):c.2044G>A (p.Ala682Thr)	EZH2	7	148809376	Weaver syndrome
NM_004456.4(EZH2):c.2233G>A (p.Glu745Lys)	EZH2	7	148807669	Weaver syndrome
NM_004456.4(EZH2):c.2050C>T (p.Arg684Cys)	EZH2	7	148809370	Weaver syndrome
NM_004456.4(EZH2):c.1876G>A (p.Val626Met)	EZH2	7	148811696	Weaver syndrome
NM_030957.3(ADAMTS10):c.709C>T (p.Arg237Ter)	ADAMTS10	19	8601029	Weill-Marchesani syndrome 1
NM_030957.3(ADAMTS10):c.1190+1G>A	ADAMTS10	19	8596306	Weill-Marchesani syndrome 1
NM_030957.3(ADAMTS10):c.810+1G>A	ADAMTS10	19	8609927	Weill-Marchesani syndrome 1
NM_030957.3(ADAMTS10):c.73G>A (p.Ala25Thr)	ADAMTS10	19	8605638	Weill-Marchesani syndrome 1
NM_030957.3(ADAMTS10):c.952C>T (p.Gln318Ter)	ADAMTS10	19	8597075	Weill-Marchesani syndrome 1
NM_030957.3(ADAMTS10):c.1553G>A (p.Gly518Asp)	ADAMTS10	19	8592797	Weill-Marchesani syndrome 1
NM_000428.2(LTBP2):c.3529G>A (p.Val1177Met)	LTBP2	14	74508727	Weill-Marchesani syndrome 1;Weill-Marchesani syndrome 3
NM_139057.3(ADAMTS17):c.760C>T (p.Gln254Ter)	ADAMTS17	15	100281258	Weill-Marchesani-like syndrome
NM_080680.2(COL11A2):c.4322G>A (p.Gly1441Glu)	COL11A2	6	33166736	Weissenbacher-Zweymuller syndrome
NM_080680.2(COL11A2):c.3991C>T (p.Arg1331Ter)	COL11A2	6	33167822	Weissenbacher-Zweymuller syndrome, autosomal recessive
NM_002180.2(IGHMBP2):c.121C>T (p.Gln41Ter)	IGHMBP2	11	68906103	Werdnig-Hoffmann disease
NM_000553.4(WRN):c.3913C>T (p.Arg1305Ter)	WRN	8	31157461	Werner syndrome
NM_000553.4(WRN):c.3493C>T (p.Gln1165Ter)	WRN	8	31147397	Werner syndrome
NM_000553.4(WRN):c.1105C>T (p.Arg369Ter)	WRN	8	31081132	Werner syndrome
NM_000553.4(WRN):c.2665C>T (p.Arg889Ter)	WRN	8	31124556	Werner syndrome
NM_000553.4(WRN):c.673C>T (p.Arg225Ter)	WRN	8	31068276	Werner syndrome
NM_006005.3(WFS1):c.2492G>A (p.Gly831Asp)	WFS1	4	6302287	WFS1-Related Disorders
NM_002272.3(KRT4):c.1303G>A (p.Glu435Lys)	KRT4	12	52807687	White sponge nevus of cannon
NM_015100.3(POGZ):c.2935C>T (p.Arg979Ter)	POGZ	1	151406100	White-sutton syndrome
NM_015100.3(POGZ):c.2590C>T (p.Arg864Ter)	POGZ	1	151406445	White-sutton syndrome
NM_015100.3(POGZ):c.3847C>T (p.Gln1283Ter)	POGZ	1	151405188	White-sutton syndrome
NM_015100.3(POGZ):c.3001C>T (p.Arg1001Ter)	POGZ	1	151406034	White-sutton syndrome;not provided
NM_018684.3(ZC4H2):c.593G>A (p.Arg198Gln)	ZC4H2	X	64917865	Wieacker syndrome

NM_018684.3(ZC4H2):c.601C>T (p.Pro201Ser)	ZC4H2	X	64917857	Wieacker syndrome	
NM_018684.3(ZC4H2):c.637C>T (p.Arg213Trp)	ZC4H2	X	64917821	Wieacker syndrome	
NM_018684.3(ZC4H2):c.225+5G>A	ZC4H2	X	64921812	Wieacker syndrome	
NM_018684.3(ZC4H2):c.53G>A (p.Arg18Lys)	ZC4H2	X	64976325	Wieacker syndrome	
NM_001197104.1(KMT2A):c.7144C>T (p.Arg2382Ter)	KMT2A	11	118503036	Wiedemann-Steiner syndrome	
NM_001197104.1(KMT2A):c.8095C>T (p.Arg2699Ter)	KMT2A	11	118503987	Wiedemann-Steiner syndrome	
NM_006306.3(SMC1A):c.121C>T (p.Leu41Phe)	SMC1A	X	53415158	Wiedemann-Steiner syndrome	
NM_001197104.1(KMT2A):c.4696+1G>A	KMT2A	11	118490250	Wiedemann-Steiner syndrome	
NM_004484.3(GPC3):c.361C>T (p.His121Tyr)	GPC3	X	133754153	Wilms tumor 1	
NM_004484.3(GPC3):c.1705G>A (p.Ala569Thr)	GPC3	X	133536162	Wilms tumor 1	
NM_000053.3(ATP7B):c.3796G>A (p.Gly1266Arg)	ATP7B	13	51937583	Wilson disease	
NM_000053.3(ATP7B):c.2621C>T (p.Ala874Val)	ATP7B	13	51950116	Wilson disease	
NM_000053.3(ATP7B):c.2293G>A (p.Asp765Asn)	ATP7B	13	51958373	Wilson disease	
NM_000053.3(ATP7B):c.2827G>A (p.Gly943Ser)	ATP7B	13	51949700	Wilson disease	
NM_000053.3(ATP7B):c.2071G>A (p.Gly691Arg)	ATP7B	13	51960198	Wilson disease	
NM_000053.3(ATP7B):c.3955C>T (p.Arg1319Ter)	ATP7B	13	51937342	Wilson disease	
NM_000053.3(ATP7B):c.2128G>A (p.Gly710Ser)	ATP7B	13	51958538	Wilson disease	
NM_000053.3(ATP7B):c.2336G>A (p.Trp779Ter)	ATP7B	13	51958330	Wilson disease	
NM_000053.3(ATP7B):c.2865+1G>A	ATP7B	13	51949661	Wilson disease	
NM_000053.3(ATP7B):c.4021G>A (p.Gly1341Ser)	ATP7B	13	51937276	Wilson disease	
NM_000053.3(ATP7B):c.3182G>A (p.Gly1061Glu)	ATP7B	13	51944170	Wilson disease	
NM_000053.3(ATP7B):c.4114C>T (p.Gln1372Ter)	ATP7B	13	51935603	Wilson disease	
NM_000053.3(ATP7B):c.1708-1G>A	ATP7B	13	51965034	Wilson disease	
NM_000053.3(ATP7B):c.2906G>A (p.Arg969Gln)	ATP7B	13	51946438	Wilson disease;not provided	
NM_000053.3(ATP7B):c.865C>T (p.Gln289Ter)	ATP7B	13	51974355	Wilson disease;not provided	
NM_000053.3(ATP7B):c.2930C>T (p.Thr977Met)	ATP7B	13	51946414	Wilson disease;not provided	
NM_000053.3(ATP7B):c.3659C>T (p.Thr1220Met)	ATP7B	13	51939091	Wilson disease;not provided	
NM_000053.3(ATP7B):c.2605G>A (p.Gly869Arg)	ATP7B	13	51950132	Wilson disease;not provided	
NM_000053.3(ATP7B):c.2975C>T (p.Pro992Leu)	ATP7B	13	51946369	Wilson disease;not provided	
NM_000053.3(ATP7B):c.2519C>T (p.Pro840Leu)	ATP7B	13	51950328	Wilson disease;not provided	
NM_000377.2(WAS):c.100C>T (p.Arg34Ter)	WAS	X	48683953	Wiskott-Aldrich syndrome	
NM_000377.2(WAS):c.37C>T (p.Arg13Ter)	WAS	X	48683890	Wiskott-Aldrich syndrome	
NM_000377.2(WAS):c.257G>A (p.Arg86His)	WAS	X	48684407	Wiskott-Aldrich syndrome;not provided	
NM_001145357.1(SIN3A):c.3310C>T (p.Arg1104Ter)	SIN3A	15	75380702	Witteveen-kolk syndrome	
NM_004836.6(EIF2AK3):c.1763G>A (p.Arg588Gln)	EIF2AK3	2	88583430	Wolcott-Rallison dysplasia	
NM_004836.6(EIF2AK3):c.2985+1G>A	EIF2AK3	2	88570873	Wolcott-Rallison dysplasia	
NM_004836.6(EIF2AK3):c.1192C>T (p.Gln398Ter)	EIF2AK3	2	88588875	Wolcott-Rallison dysplasia	
NM_006005.3(WFS1):c.2338G>A (p.Gly780Ser)	WFS1	4	6302133	Wolfram-like syndrome, autosomal dominant	
NM_002335.3(LRP5):c.640G>A (p.Ala214Thr)	LRP5	11	68357801	Worth disease	
NM_002335.3(LRP5):c.641C>T (p.Ala214Val)	LRP5	11	68357802	Worth disease	
NM_000039.2(APOA1):c.166C>T (p.Gln56Ter)	APOA1	11	116837035	Xanthelasmas, periorbital	
NM_017947.3(MOCOS):c.1255C>T (p.Arg419Ter)	MOCOS	18	36213402	Xanthinuria type 2	
NM_017947.3(MOCOS):c.2326C>T (p.Arg776Cys)	MOCOS	18	36260092	Xanthinuria type 2	
NM_001142864.3(PIEZO1):c.7367G>A (p.Arg2456His)	PIEZO1	16	88715804	Xerocytosis	
NM_001142864.3(PIEZO1):c.6380C>T (p.Thr2127Met)	PIEZO1	16	88719665	Xerocytosis	

NM_001142864.3(PIEZO1):c.6058G>A (p.Ala2020Thr)	PIEZO1	16	88720175	Xerocytosis;not provided
NM_000123.3(ERCC5):c.787C>T (p.Arg263Ter)	ERCC5	13	102861621	Xeroderma pigmentosum group g/Cockayne syndrome
NM_000123.3(ERCC5):c.526C>T (p.Gln176Ter)	ERCC5	13	102856110	Xeroderma pigmentosum group g/Cockayne syndrome
NM_000122.1(ERCC3):c.1273C>T (p.Arg425Ter)	ERCC3	2	127286772	Xeroderma pigmentosum, complementation group b
NM_000122.1(ERCC3):c.1633C>T (p.Gln545Ter)	ERCC3	2	127279270	Xeroderma pigmentosum, complementation group b
NM_004628.4(XPC):c.1735C>T (p.Arg579Ter)	XPC	3	14158148	Xeroderma pigmentosum, group C
NM_000400.3(ERCC2):c.2176C>T (p.Gln726Ter)	ERCC2	19	45352223	Xeroderma pigmentosum, group D
NM_000400.3(ERCC2):c.2047C>T (p.Arg683Trp)	ERCC2	19	45352352	Xeroderma pigmentosum, group D
NM_000400.3(ERCC2):c.2048G>A (p.Arg683Gln)	ERCC2	19	45352351	Xeroderma pigmentosum, group D
NM_000107.2(DDB2):c.818G>A (p.Arg273His)	DDB2	11	47234872	Xeroderma pigmentosum, group E
NM_000107.2(DDB2):c.937C>T (p.Arg313Ter)	DDB2	11	47235326	Xeroderma pigmentosum, group E
NM_000123.3(ERCC5):c.2375C>T (p.Ala792Val)	ERCC5	13	102866687	Xeroderma pigmentosum, group G
NM_000123.3(ERCC5):c.406C>T (p.Gln136Ter)	ERCC5	13	102854313	Xeroderma pigmentosum, group G
NM_000380.3(XPA):c.619C>T (p.Arg207Ter)	XPA	9	97684977	Xeroderma pigmentosum, type 1
NM_000380.3(XPA):c.682C>T (p.Arg228Ter)	XPA	9	97675579	Xeroderma pigmentosum, type 1
NM_005236.2(ERCC4):c.1765C>T (p.Arg589Trp)	ERCC4	16	13935697	Xeroderma pigmentosum, type f/Cockayne syndrome
NM_006502.2(POLH):c.376C>T (p.Gln126Ter)	POLH	6	43587375	Xeroderma pigmentosum, variant type
NM_006502.2(POLH):c.1117C>T (p.Gln373Ter)	POLH	6	43610596	Xeroderma pigmentosum, variant type
NM_006502.2(POLH):c.764+1G>A	POLH	6	43601092	Xeroderma pigmentosum, variant type
NM_006502.2(POLH):c.907C>T (p.Arg303Ter)	POLH	6	43604637	Xeroderma pigmentosum, variant type
NM_000061.2(BTK):c.1574G>A (p.Arg525Gln)	BTK	X	101354687	X-linked agammaglobulinemia
NM_000061.2(BTK):c.37C>T (p.Arg13Ter)	BTK	X	101375248	X-linked agammaglobulinemia
NM_000061.2(BTK):c.43C>T (p.Gln15Ter)	BTK	X	101375242	X-linked agammaglobulinemia
NM_000061.2(BTK):c.755G>A (p.Trp252Ter)	BTK	X	101360589	X-linked agammaglobulinemia
NM_000061.2(BTK):c.763C>T (p.Arg255Ter)	BTK	X	101360581	X-linked agammaglobulinemia
NM_000061.2(BTK):c.862C>T (p.Arg288Trp)	BTK	X	101359325	X-linked agammaglobulinemia
NM_000061.2(BTK):c.1559G>A (p.Arg520Gln)	BTK	X	101356059	X-linked agammaglobulinemia
NM_000061.2(BTK):c.1684C>T (p.Arg562Trp)	BTK	X	101353936	X-linked agammaglobulinemia
NM_000061.2(BTK):c.1838G>A (p.Gly613Asp)	BTK	X	101353264	X-linked agammaglobulinemia
NM_000061.2(BTK):c.1750+5G>A	BTK	X	101353865	X-linked agammaglobulinemia with growth hormone deficiency
NM_000061.2(BTK):c.83G>A (p.Arg28His)	BTK	X	101375202	X-linked agammaglobulinemia;not provided
NM_000061.2(BTK):c.1558C>T (p.Arg520Ter)	BTK	X	101356060	X-linked agammaglobulinemia;not provided
NM_001097642.2(GJB1):c.514C>T (p.Pro172Ser)	GJB1	X	71224221	X-linked hereditary motor and sensory neuropathy
NM_000166.5(GJB1):c.415G>A (p.Val139Met)	GJB1	X	71224122	X-linked hereditary motor and sensory neuropathy
NM_000166.5(GJB1):c.556G>A (p.Glu186Lys)	GJB1	X	71224263	X-linked hereditary motor and sensory neuropathy
NM_001097642.2(GJB1):c.172C>T (p.Pro58Ser)	GJB1	X	71223879	X-linked hereditary motor and sensory neuropathy
NM_000166.5(GJB1):c.790C>T (p.Arg264Cys)	GJB1	X	71224497	X-linked hereditary motor and sensory neuropathy
NM_001097642.2(GJB1):c.224G>A (p.Arg75Gln)	GJB1	X	71223931	X-linked hereditary motor and sensory neuropathy
NM_001097642.2(GJB1):c.319C>T (p.Arg107Trp)	GJB1	X	71224026	X-linked hereditary motor and sensory neuropathy
NM_000166.5(GJB1):c.164C>T (p.Thr55Ile)	GJB1	X	71223871	X-linked hereditary motor and sensory neuropathy;not provided
NM_000166.5(GJB1):c.187G>A (p.Val63Ile)	GJB1	X	71223894	X-linked hereditary motor and sensory neuropathy;not provided
NM_000166.5(GJB1):c.223C>T (p.Arg75Trp)	GJB1	X	71223930	X-linked hereditary motor and sensory neuropathy;not provided
NM_000166.5(GJB1):c.536G>A (p.Cys179Tyr)	GJB1	X	71224243	X-linked hereditary motor and sensory neuropathy;not provided
NM_000166.5(GJB1):c.490C>T (p.Arg164Trp)	GJB1	X	71224197	X-linked hereditary motor and sensory neuropathy;not provided
NM_000425.4(L1CAM):c.791G>A (p.Cys264Tyr)	L1CAM	X	153870403	X-linked hydrocephalus syndrome

NM_000425.4(L1CAM):c.551G>A (p.Arg184Gln)	L1CAM	X	153870933	X-linked hydrocephalus syndrome
NM_000425.4(L1CAM):c.924C>T (p.Gly308=)	L1CAM	X	153870123	X-linked hydrocephalus syndrome
NM_000425.4(L1CAM):c.2380C>T (p.Gln794Ter)	L1CAM	X	153866700	X-linked hydrocephalus syndrome
NM_000425.4(L1CAM):c.1354G>A (p.Gly452Arg)	L1CAM	X	153868866	X-linked hydrocephalus syndrome;not provided
NM_000351.5(STS):c.1337G>A (p.Cys446Tyr)	STS	X	7334066	X-linked ichthyosis with steryl-sulfatase deficiency
NM_000351.5(STS):c.1022C>T (p.Ser341Leu)	STS	X	7305109	X-linked ichthyosis with steryl-sulfatase deficiency
NM_001493.2(GDI1):c.208C>T (p.Arg70Ter)	GDI1	X	154438819	X-Linked Mental Retardation 41
NM_000206.2(IL2RG):c.341G>A (p.Gly114Asp)	IL2RG	X	71110617	X-linked severe combined immunodeficiency
NM_000206.2(IL2RG):c.854G>A (p.Arg285Gln)	IL2RG	X	71108599	X-linked severe combined immunodeficiency
NM_000206.2(IL2RG):c.865C>T (p.Arg289Ter)	IL2RG	X	71108336	X-linked severe combined immunodeficiency;not provided
NM_000206.2(IL2RG):c.677G>A (p.Arg226His)	IL2RG	X	71109308	X-linked severe combined immunodeficiency;not provided
NM_000206.2(IL2RG):c.676C>T (p.Arg226Cys)	IL2RG	X	71109309	X-linked severe combined immunodeficiency;not provided
NM_000206.2(IL2RG):c.670C>T (p.Arg224Trp)	IL2RG	X	71109315	X-linked severe combined immunodeficiency;not provided
NM_016111.3(TELO2):c.2296G>A (p.Val766Met)	TELO2	16	1507605	You-Hoover-Fong syndrome
NM_016111.3(TELO2):c.779C>T (p.Pro260Leu)	TELO2	16	1497457	You-Hoover-Fong syndrome
NM_016111.3(TELO2):c.1826G>A (p.Arg609His)	TELO2	16	1502986	You-Hoover-Fong syndrome;not provided
NM_012330.3(KAT6B):c.3664+1G>A	KAT6B	10	75025250	Young Simpson syndrome
NM_012330.3(KAT6B):c.5389C>T (p.Arg1797Ter)	KAT6B	10	75030213	Young Simpson syndrome;not provided
NM_014845.5(FIG4):c.311G>A (p.Gly104Asp)	FIG4	6	109727130	Yunis Varon syndrome
NM_000466.2(PEX1):c.2926+1G>A	PEX1	7	92494486	Zellweger syndrome
NM_000466.2(PEX1):c.2383C>T (p.Arg795Ter)	PEX1	7	92501923	Zellweger syndrome
NM_002238.3(KCNH1):c.1405G>A (p.Gly469Arg)	KCNH1	1	210804143	Zimmermann-Laband syndrome 1
NM_002238.3(KCNH1):c.1042G>A (p.Gly348Arg)	KCNH1	1	210919979	Zimmermann-Laband syndrome 1
NM_021954.3(GJA3):c.560C>T (p.Pro187Leu)	GJA3	13	20142729	Zonular pulverulent cataract 3
NM_021954.3(GJA3):c.227G>A (p.Arg76His)	GJA3	13	20143062	Zonular pulverulent cataract 3
NM_021954.3(GJA3):c.5G>A (p.Gly2Asp)	GJA3	13	20143284	Zonular pulverulent cataract 3
NM_021954.3(GJA3):c.427G>A (p.Gly143Arg)	GJA3	13	20142862	Zonular pulverulent cataract 3
NM_138927.2(SON):c.286C>T (p.Gln96Ter)	SON	21	33549517	ZTTK SYNDROME
NM_004278.3(PIGL):c.652C>T (p.Gln218Ter)	PIGL	17	16317900	Zunich neuroectodermal syndrome

[001006] ADDITIONAL EMBODIMENTS

[001007] Embodiment 1. A method of modifying an Adenine in a target locus of interest, comprising delivering to said locus: (a) a Cpf1 nickase protein; (b) a guide molecule which comprises a guide sequence linked to a direct repeat sequence; and (c) an adenosine deaminase protein or catalytic domain thereof; wherein said adenosine deaminase protein or catalytic domain thereof is covalently or non-covalently linked to said Cpf1 nickase protein or said guide molecule or is adapted to link thereto after delivery; wherein guide molecule forms a complex with said Cpf1 nickase protein and directs said complex to bind a first DNA strand at said target locus of interest, wherein said guide sequence is capable of hybridizing with a target sequence comprising said Adenine within said first DNA strand to form a heteroduplex, wherein said guide sequence comprises a non-pairing Cytosine at a position corresponding to said Adenine resulting in an A-C mismatch in the heteroduplex formed; wherein said Cpf1 nickase protein nicks a second DNA strand at said target locus of interest displaced by formation of said heteroduplex; and wherein said adenosine deaminase protein or catalytic domain thereof deaminates said Adenine in said heteroduplex.

[001008] Embodiment 2. The method of Embodiment 1, wherein said adenosine deaminase protein or catalytic domain thereof is fused to N- or C-terminus of said Cpf1 nickase protein.

[001009] Embodiment 3. The method of Embodiment 2, wherein said adenosine deaminase protein or catalytic domain thereof is fused to said Cpf1 nickase protein by a linker.

[001010] Embodiment 4. The method of Embodiment 3, wherein said linker is (GGGGS)3-11 (SEQ ID NOS:1-9), GSG5 (SEQ ID NO:10) or LEPGEKPYKCPECGKSFSQSGALTRHQRTHTR (SEQ ID NO:11).

[001011] Embodiment 5. The method of Embodiment 1, wherein said adenosine deaminase protein or catalytic domain thereof is linked to an adaptor protein and said guide molecule or said Cpf1 nickase protein comprises an aptamer sequence capable of binding to said adaptor protein.

[001012] Embodiment 6. The method of Embodiment 5, wherein said adaptor sequence is selected from MS2, PP7, Qβ, F2, GA, fr, JP501, M12, R17, BZ13, JP34, JP500, KU1, M11,

MX1, TW18, VK, SP, FI, ID2, NL95, TW19, AP205, ϕCb5, ϕCb8r, ϕCb12r, ϕCb23r, 7s and PRR1.

[001013] Embodiment 7. The method of Embodiment 1, wherein said adenosine deaminase protein or catalytic domain thereof is inserted into an internal loop of said Cpf1 nickase protein.

[001014] Embodiment 8. The method of any of Embodiments 1-7, wherein said Cpf1 nickase protein comprises a mutation in the Nuc domain.

[001015] Embodiment 9. The method of Embodiment 8, wherein said Cpf1 nickase protein comprises a mutation corresponding to R1226A in AsCpf1.

[001016] Embodiment 10. The method of any of Embodiments 1-7, wherein said Cpf1 nickase protein has at least part of the Nuc domain removed.

[001017] Embodiment 11. The method of any of Embodiments 1-10, wherein said guide molecule binds to said Cpf1 nickase protein and is capable of forming said heteroduplex of about 24 nt with said target sequence.

[001018] Embodiment 12. The method of any of Embodiments 1-10, wherein said guide molecule binds to said Cpf1 nickase protein and is capable of forming said heteroduplex of more than 24 nt with said target sequence.

[001019] Embodiment 13. The method of any of the preceding Embodiments, wherein said adenosine deaminase protein or catalytic domain thereof is a human, squid or Drosophila adenosine deaminase protein or catalytic domain thereof.

[001020] Embodiment 14. The method of Embodiment 13, wherein said adenosine deaminase protein or catalytic domain thereof has been modified to increase activity against a DNA-RNA heteroduplex .

[001021] Embodiment 15. The method of Embodiment 14, wherein said adenosine deaminase protein or catalytic domain thereof is a mutated hADAR2d comprising mutation E488Q or a mutated hADAR1d comprising mutation E1008Q.

[001022] Embodiment 16. The method of Embodiment 13, wherein said adenosine deaminase protein or catalytic domain thereof has been modified to reduce off-target effects.

[001023] Embodiment 17. The method of Embodiment 16, wherein said adenosine deaminase protein or catalytic domain thereof is a mutated hADAR2d comprising mutation T375G/S, N473D, or both, or a mutated hADAR1d comprising corresponding mutations.

[001024] Embodiment 18. The method of any of the preceding Embodiments, wherein said Cpf1 nickase protein and optionally said adenosine deaminase protein or catalytic domain thereof comprise one or more heterologous nuclear localization signal(s) (NLS(s)).

[001025] Embodiment 19. The method of any of the preceding Embodiments, wherein said method comprises, determining said target sequence of interest and selecting said adenosine deaminase protein or catalytic domain thereof which most efficiently deaminates said Adenine present in said target sequence.

[001026] Embodiment 20. The method of any of the preceding Embodiments, wherein said Cpf1 nickase protein is obtained from a Cpf1 nuclease derived from a bacterial species selected from the group consisting of Francisella tularensis, Prevotella albensis, Lachnospiraceae bacterium, Butyrivibrio proteoclasticus, Peregrinibacteria bacterium, Parcubacteria bacterium, Smithella sp., Acidaminococcus sp., Lachnospiraceae bacterium, Candidatus Methanoplasma termitum, Eubacterium eligens, Moraxella bovoculi, Leptospira inadai, Porphyromonas crevioricanis, Prevotella disiens and Porphyromonas macacae, Succinivibrio dextrinosolvens, Prevotella disiens, Flavobacterium branchiophilum, Helcococcus kunzii, Eubacterium sp., Microgenomates (Roizmanbacteria) bacterium, Flavobacterium sp., Prevotella brevis, Moraxella caprae, Bacteroidetes oral, Porphyromonas cansulci, Synergistes jonesii, Prevotella bryantii, Anaerovibrio sp., Butyrivibrio fibrisolvens, Candidatus Methanomethylophilus, Butyrivibrio sp., Oribacterium sp., Pseudobutyrivibrio ruminis and Proteocatella sphenisci.

[001027] Embodiment 21. The method of Embodiment 20, wherein said Cpf1 nickase protein is a FnCpf1 nickase and recognizes a PAM sequence of TTN, wherein N is A/C/G or T, or said Cpf1 nickase protein is a PaCpf1p, LbCpf1 or AsCpf1 nickase and recognizes a PAM sequence of TTTV, wherein V is A/C or G.

[001028] Embodiment 22. The method of Embodiment 20, wherein said Cpf1 nickase protein has been modified and recognizes an altered PAM sequence.

[001029] Embodiment 23. The method of any of the preceding Embodiments, wherein said target locus of interest is within a cell.

[001030] Embodiment 24. The method of Embodiment 23, wherein said cell is a eukaryotic cell.

[001031] Embodiment 25. The method of Embodiment 23, wherein said cell is a non-human animal cell.

[001032] Embodiment 26. The method of Embodiment 23, wherein said cell is a human cell.

[001033] Embodiment 27. The method of Embodiment 23, wherein said cell is a plant cell.

[001034] Embodiment 28. The method of any of the preceding Embodiments, wherein said target locus of interest is within an animal.

[001035] Embodiment 29. The method of any of the preceding Embodiments, wherein said target locus of interest is within a plant.

[001036] Embodiment 30. The method of any of the preceding Embodiments, wherein said target locus of interest is comprised in a DNA molecule in vitro.

[001037] Embodiment 31. The method of any of the preceding Embodiments, wherein said components (a), (b) and (c) are delivered to said cell as a ribonucleoprotein complex.

[001038] Embodiment 32. The method of any of the preceding Embodiments, wherein said components (a), (b) and (c) are delivered to said cell as one or more polynucleotide molecules.

[001039] Embodiment 33. The method of Embodiment 32, wherein said one or more polynucleotide molecules comprise one or more mRNA molecules encoding components (a) and/or (c).

[001040] Embodiment 34. The method of Embodiment 32, wherein said one or more polynucleotide molecules are comprised within one or more vectors.

[001041] Embodiment 35. The method of Embodiment 34, wherein said one or more polynucleotide molecules comprise one or more regulatory elements operably configured to express said Cpf1 nickase protein, said guide molecule, and said adenosine deaminase protein or catalytic domain thereof, optionally wherein said one or more regulatory elements comprise inducible promoters.

[001042] Embodiment 36. The method of any of Embodiments 31-35, wherein said one or more polynucleotide molecules or said ribonucleoprotein complex are delivered via particles, vesicles, or one or more viral vectors.

[001043] Embodiment 37. The method of Embodiment 36, wherein said particles comprise a lipid, a sugar, a metal or a protein.

[001044] Embodiment 38. The method of Embodiment 37, wherein said particles comprise lipid nanoparticles.

[001045] Embodiment 39. The method of Embodiment 36, wherein said vesicles comprise exosomes or liposomes.

[001046] Embodiment 40. The method of Embodiment 36, wherein said one or more viral vectors comprise one or more of adenovirus, one or more lentivirus or one or more adeno-associated virus.

[001047] Embodiment 41. The method of any of the preceding Embodiments, where said method modifies a cell, a cell line or an organism by manipulation of one or more target sequences at genomic loci of interest.

[001048] Embodiment 42. The method of Embodiment 41, wherein said deamination of said Adenine at said target locus of interest remedies a disease caused by a G→A or C→T point mutation or a pathogenic SNP.

[001049] Embodiment 43. The method of Embodiment 42, wherein said disease is selected from cancer, haemophilia, beta-thalassemia, Marfan syndrome and Wiskott-Aldrich syndrome.

[001050] Embodiment 44. The method of Embodiment 41, wherein said deamination of said Adenine at said target locus of interest inactivates a target gene at said target locus.

[001051] Embodiment 45. A modified cell obtained from the method of any of the preceding Embodiments, or progeny of said modified cell, wherein said cell comprises a hypoxanthine or a guanine in replace of said Adenine in said target locus of interest compared to a corresponding cell not subjected to the method.

[001052] Embodiment 46. The modified cell or progeny thereof of Embodiment 45, wherein said cell is a eukaryotic cell.

[001053] Embodiment 47. The modified cell or progeny thereof of Embodiment 45, wherein said cell is an animal cell.

[001054] Embodiment 48. The modified cell or progeny thereof of Embodiment 45, wherein said cell is a human cell.

[001055] Embodiment 49. The modified cell or progeny thereof of Embodiment 45, wherein said cell is a therapeutic T cell.

[001056] Embodiment 50. The modified cell or progeny thereof of Embodiment 45, wherein said cell is an antibody-producing B cell.

[001057] Embodiment 51. The modified cell or progeny thereof of Embodiment 45, wherein said cell is a plant cell.

[001058] Embodiment 52. A non-human animal comprising said modified cell of Embodiment 47.

[001059] Embodiment 53. A plant comprising said modified cell of Embodiment 51.

[001060] Embodiment 54. A method for cell therapy, comprising administering to a patient in need thereof said modified cell of any of Embodiments 45-50, wherein presence of said modified cell remedies a disease in the patient.

[001061] Embodiment 55. An engineered, non-naturally occurring system suitable for modifying an Adenine in a target locus of interest, comprising: a guide molecule which comprises a guide sequence linked to a direct repeat sequence, or a nucleotide sequence encoding said guide molecule; a Cpf1 nickase protein, or a nucleotide sequence encoding said Cpf1 nickase protein; an adenosine deaminase protein or catalytic domain thereof, or a nucleotide sequence encoding said adenosine deaminase protein or catalytic domain thereof; wherein said adenosine deaminase protein or catalytic domain thereof is covalently or non-covalently linked to said Cpf1 nickase protein or said guide molecule or is adapted to link thereto after delivery; wherein said guide sequence is capable of hybridizing with a target sequence comprising an Adenine on a first DNA strand at said target locus to form a heteroduplex, wherein said guide sequence comprises a non-pairing Cytosine at a position corresponding to said Adenine resulting in an A-C mismatch in the heteroduplex formed; and wherein said Cpf1 nickase protein is capable of nicking a second DNA strand complementary to said first DNA strand.

[001062] Embodiment 56. An engineered, non-naturally occurring vector system suitable for modifying an Adenine in a target locus of interest, comprising the nucleotide sequences of a), b) and c) of Embodiment 55.

[001063] Embodiment 57. The engineered, non-naturally occurring vector system of Embodiment 56, comprising one or more vectors comprising: a first regulatory element operably linked to a nucleotide sequence encoding said guide molecule which comprises said guide sequence; a second regulatory element operably linked to a nucleotide sequence encoding said Cpf1 nickase protein; and a nucleotide sequence encoding an adenosine deaminase protein or catalytic domain thereof which is under control of said first or second regulatory element or operably linked to a third regulatory element; wherein, if said nucleotide sequence encoding an adenosine deaminase protein or catalytic domain thereof is operably linked to a third regulatory element, said adenosine deaminase protein or catalytic domain thereof is adapted to link to said guide molecule or said Cpf1 nickase protein after expression; wherein components (a), (b) and (c) are located on the same or different vectors of the system.

[001064] Embodiment 58. An in vitro or ex vivo host cell or progeny thereof or cell line or progeny thereof comprising the system of any of Embodiments 55-57.

[001065] Embodiment 59. The host cell or progeny thereof or cell line or progeny thereof of Embodiment 58, wherein said cell is a eukaryotic cell.

[001066] Embodiment 60. The host cell or progeny thereof or cell line or progeny thereof of Embodiment 58, wherein said cell is an animal cell.

[001067] Embodiment 61. The host cell or progeny thereof or cell line or progeny thereof of Embodiment 58, wherein said cell is a human cell.

[001068] Embodiment 62. The host cell or progeny thereof or cell line or progeny thereof of Embodiment 58, wherein said cell is a plant cell.

WORKING EXAMPLES

[001069] EXAMPLE 1

[001070] Adenine deaminases (ADs) typically deaminates adenines at specific sites in double stranded RNA. Previous efforts have attempted to evolve ADs to change their substrate preference from dsRNA to dsDNA, so that the evolved AD can be fused to Cpf1 to achieve RNA-guided adenine deamination on genomic DNA.

[001071] The facts that some ADs can effect adenine deamination on DNA-RNA heteroduplexes (e.g. Zheng et al., Nucleic Acids Research 2017) presents a unique opportunity to develop an RNA guided AD by taking advantage of the heteroduplex formed between the guide RNA and its complementary DNA target in the R-loop formed during RNA-guided DNA binding by inactive Cpf1. By using inactive Cpf1 to recruit an AD, the AD enzyme will then act on the adenine in the RNA-DNA heteroduplex.

[001072] In one embodiment, an inactive AsCpf1 is obtained using the following mutations: D908A or E993A. To increase the efficiency of editing by AD, nickase Cpf1 can also be used to nick the strand of DNA that is not complementary to the guide RNA. For AsCpf1 the mutation would be R1226A.

[001073] Designs for the recruitment of AD to a specific locus:

[001074] 1. NLS-tagged inactive or nickase Cpf1 is fused to AD on either the N- or C-terminal end. A variety of linkers are used including flexible linkers such as GSG_5 or less flexible linkers such as LEPGEKPYKCPECGKSFSQSGALTRHQRTHTR (SEQ ID NO:78).

[001075] 2. The guide RNA scaffold is modified with aptamers such as MS2 binding sites (e.g. Konermann et al., Nature 2015). NLS-tagged AD-MS2 binding protein fusions is co-introduced into target cells along with (NLS-tagged inactive or nickase Cpf1) and corresponding guide RNA.

[001076] 3. AD is inserted into an internal loop of NLS-tagged inactive or nickase Cpf1.

[001077] Designs for the RNA guide:

[001078] 1. Normal length of RNA guide (24nt for AsCpf1) is designed to target the genomic locus of interest.

[001079] 2. RNA guide with longer than canonical length is used to form heteroduplexes outside of the protein-guide RNA-target DNA complex.

[001080] For each of these RNA guide designs, the base on the RNA that is opposite of the adenine on the DNA strand would be specified as a C as opposed to U.

[001081] Choice and Designs of ADs:

[001082] A number of ADs are used, and each will have varying levels of activity. These ADs include:

[001083] 1. Human ADARs (hADAR1, hADAR2, hADAR3)

[001084] 2. Squid *Loligo pealeii* ADARs (sqADAR2a, sqADAR2b)

[001085] 3. ADATs (human ADAT, *Drosophila* ADAT)

[001086] Mutations can also be used to increase the activity of ADAR reacting against a DNA-RNA heteroduplex. For example, for the human ADAR genes, the hADAR1d(E1008Q) or hADAR2d(E488Q) mutation is used to increase their activity against a DNA-RNA target.

[001087] Each ADAR has varying levels of sequence context requirement. For example, for hADAR1d (E1008Q), tAg and aAg sites are efficiently deaminated, whereas aAt and cAc are less efficiently edited, and gAa and gAc are even less edited. However, the context requirement will vary for different ADARs.

[001088] A schematic showing of one version of the system is provided in Figure 1. The amino acid sequences of example Cpf1-AD fusion proteins are provided in Figures 2 to 5.

[001089] EXAMPLE 2

[001090] Figure 6 shows SpCas9 & AsCpf1 fusions with huADAR2d. Four constructs for AsCpf1 and four Constructs for SpCas9 for A to G conversion were made. A nickase version for AsCpf1 (R1226A) and SpCas9 (N863A) was fused at the N or C-term with the deaminase domain of human ADAR2 (ADAR). Additionally, deletion constructs were generated by removing the HNH domain from SpCas9 or the Nuc domain from AsCpf1 to reduce steric hindrance for ADAR.

[001091] Figure 7 shows deletion constructs for ADAR fusions. Amino acids 1076 to 1258 of AsCpf1 were replaced with a GSGG linker, and amino acids 769 to 918 of SpCas9 were replaced with a GGSGGS linker.

[001092] Figure 8 shows expression of ADAR fusions in HEK cells. HEK293T cells were transfected with different ADAR fusion constructs or HNH/Nuc deletion constructs to confirm protein expression. Cells were harvested two days after transfection and protein was extracted using RIPA buffer. 5ul of cell lysate was used for western blot, using antibodies against a Flag (SpCas9) or HA (AsCpf1) tag.

[001093] HEK293T cells were transfected with three construct. One provides a luciferase (Cluc) mRNA target that has a UAG motif at a defined position (Figure 9, left). The TAG motif was tiled over a 20nt region, resulting in 10 targets which are potentially more or less

accessible for the ADAR2d (SEQ ID NOS:85-94). For each of the 10 target constructs a matching crRNA or sgRNA, which contains the programed A to G conversion, was provided (Figure 9, right) (SEQ ID NOS:95-104).

[001094] Cells were harvested using the cells to CT protocol (Joung et al., Nat Protoc. 2017). cDNA was amplified by PCR and sequences by NGS. Three to six technical replicates where performed for each ADAR fusion construct and guide/target combination.

[001095] AsCpf1 with C-terminal ADAR fusions performed the best with up to ~30% A to G conversion at nucleotide position 18 (target/guide #2), counted from were the PAM would be on a DNA target (Figure 10, left). In constrast, SpCas9 ADAR fusion did not result in A to G conversion above the background observed with the ADAR only control (Figure 10, right).

[001096] In addition, targeted A-to-G DNA base editing of human DNMT1 gene by AsCpf1-ADAR2d constructs were tested in HEK293 cells. As shown in Figure 11, AsCpf1(R1226A)-ADAR2d and AsCpf1(deltaNuc)-ADAR2d fusion constructs, in complex with gRNA targeting human DNMT1 gene, each exhibited a detectable level of targeted A-to-G DNA base editing, whereas wtAsCpf1 and ADAR2d control constructs did not result in a detectable level of targeted A-to-G DNA base editing.

[001097] The embodiments, illustratively described herein may suitably be practiced in the absence of any element or elements, limitation or limitations, not specifically disclosed herein. Thus, for example, the terms "comprising," "including," "containing," etc. shall be read expansively and without limitation. Additionally, the terms and expressions employed herein have been used as terms of description and not of limitation, and there is no intention in the use of such terms and expressions of excluding any equivalents of the features shown and described or portions thereof, but it is recognized that various modifications are possible within the scope of the claimed technology. Additionally, the phrase "consisting essentially of" will be understood to include those elements specifically recited and those additional elements that do not materially affect the basic and novel characteristics of the claimed technology. The phrase "consisting of" excludes any element not specified.

[001098] The present disclosure is not to be limited in terms of the particular embodiments described in this application. Many modifications and variations can be made without departing from its spirit and scope, as will be apparent to those skilled in the art. Functionally equivalent methods and compositions within the scope of the disclosure, in

addition to those enumerated herein, will be apparent to those skilled in the art from the foregoing descriptions. Such modifications and variations are intended to fall within the scope of the appended claims. The present disclosure is to be limited only by the terms of the appended claims, along with the full scope of equivalents to which such claims are entitled. It is to be understood that this disclosure is not limited to particular methods, reagents, compounds compositions or biological systems, which can of course vary. It is also to be understood that the terminology used herein is for the purpose of describing particular embodiments only, and is not intended to be limiting.

[001099] In addition, where features or aspects of the disclosure are described in terms of Markush groups, those skilled in the art will recognize that the disclosure is also thereby described in terms of any individual member or subgroup of members of the Markush group.

[001100] As will be understood by one skilled in the art, for any and all purposes, particularly in terms of providing a written description, all ranges disclosed herein also encompass any and all possible subranges and combinations of subranges thereof. Any listed range can be easily recognized as sufficiently describing and enabling the same range being broken down into at least equal halves, thirds, quarters, fifths, tenths, etc. As a non-limiting example, each range discussed herein can be readily broken down into a lower third, middle third and upper third, etc. As will also be understood by one skilled in the art all language such as "up to," "at least," "greater than," "less than," and the like, include the number recited and refer to ranges which can be subsequently broken down into subranges as discussed above. Finally, as will be understood by one skilled in the art, a range includes each individual member.

[001101] All publications, patent applications, issued patents, and other documents referred to in this specification are herein incorporated by reference as if each individual publication, patent application, issued patent, or other document was specifically and individually indicated to be incorporated by reference in its entirety. Definitions that are contained in text incorporated by reference are excluded to the extent that they contradict definitions in this disclosure.

[001102] Other embodiments are set forth in the following claims.

WHAT IS CLAIMED IS:

1. A method of modifying an Adenine in a target locus of interest, comprising delivering to said locus:

(a) a Cpf1 nickase protein;

(b) a guide molecule which comprises a guide sequence linked to a direct repeat sequence; and

(c) an adenosine deaminase protein or catalytic domain thereof;

wherein said adenosine deaminase protein or catalytic domain thereof is covalently or non-covalently linked to said Cpf1 nickase protein or said guide molecule or is adapted to link thereto after delivery;

wherein guide molecule forms a complex with said Cpf1 nickase protein and directs said complex to bind a first DNA strand at said target locus of interest, wherein said guide sequence is capable of hybridizing with a target sequence comprising said Adenine within said first DNA strand to form a heteroduplex, wherein said guide sequence comprises a non-pairing Cytosine at a position corresponding to said Adenine resulting in an A-C mismatch in the heteroduplex formed;

wherein said Cpf1 nickase protein nicks a second DNA strand at said target locus of interest displaced by formation of said heteroduplex; and

wherein said adenosine deaminase protein or catalytic domain thereof deaminates said Adenine in said heteroduplex.

2. The method of claim 1, wherein said adenosine deaminase protein or catalytic domain thereof is fused to N- or C-terminus of said Cpf1 nickase protein.

3. The method of claim 2, wherein said adenosine deaminase protein or catalytic domain thereof is fused to said Cpf1 nickase protein by a linker.

4. The method of claim 3, wherein said linker is $(GGGGS)_{3-11}$ (SEQ ID NOS:1-9), GSG_5 (SEQ ID NO:10) or LEPGEKPYKCPECGKSFSQSGALTRHQRTHTR (SEQ ID NO:11).

5. The method of claim 1, wherein said adenosine deaminase protein or catalytic domain thereof is linked to an adaptor protein and said guide molecule or said Cpf1 nickase protein comprises an aptamer sequence capable of binding to said adaptor protein.

6. The method of claim 5, wherein said adaptor sequence is selected from MS2, PP7, Qβ, F2, GA, fr, JP501, M12, R17, BZ13, JP34, JP500, KU1, M11, MX1, TW18, VK, SP, FI, ID2, NL95, TW19, AP205, ϕCb5, ϕCb8r, ϕCb12r, ϕCb23r, 7s and PRR1.

7. The method of claim 1, wherein said adenosine deaminase protein or catalytic domain thereof is inserted into an internal loop of said Cpf1 nickase protein.

8. The method of any of claims 1-7, wherein said Cpf1 nickase protein comprises a mutation in the Nuc domain.

9. The method of claim 8, wherein said Cpf1 nickase protein comprises a mutation corresponding to R1226A in AsCpf1.

10. The method of any of claims 1-7, wherein said Cpf1 nickase protein has at least part of the Nuc domain removed.

11. The method of any of claims 1-10, wherein said guide molecule binds to said Cpf1 nickase protein and is capable of forming said heteroduplex of about 24 nt with said target sequence.

12. The method of any of claims 1-10, wherein said guide molecule binds to said Cpf1 nickase protein and is capable of forming said heteroduplex of more than 24 nt with said target sequence.

13. The method of any of the preceding claims, wherein said adenosine deaminase protein or catalytic domain thereof is a human, squid or Drosophila adenosine deaminase protein or catalytic domain thereof.

14. The method of claim 13, wherein said adenosine deaminase protein or catalytic domain thereof has been modified to increase activity against a DNA-RNA heteroduplex.

15. The method of claim 14, wherein said adenosine deaminase protein or catalytic domain thereof is a mutated hADAR2d comprising mutation E488Q or a mutated hADAR1d comprising mutation E1008Q.

16. The method of claim 13, wherein said adenosine deaminase protein or catalytic domain thereof has been modified to reduce off-target effects.

17. The method of claim 16, wherein said adenosine deaminase protein or catalytic domain thereof is a mutated hADAR2d comprising mutation T375G/S, N473D, or both, or a mutated hADAR1d comprising corresponding mutations.

18. The method of any of the preceding claims, wherein said Cpf1 nickase protein and optionally said adenosine deaminase protein or catalytic domain thereof comprise one or more heterologous nuclear localization signal(s) (NLS(s)).

19. The method of any of the preceding claims, wherein said method comprises, determining said target sequence of interest and selecting said adenosine deaminase protein or catalytic domain thereof which most efficiently deaminates said Adenine present in said target sequence.

20. The method of any of the preceding claims, wherein said Cpf1 nickase protein is obtained from a Cpf1 nuclease derived from a bacterial species selected from the group consisting of Francisella tularensis, Prevotella albensis, Lachnospiraceae bacterium, Butyrivibrio proteoclasticus, Peregrinibacteria bacterium, Parcubacteria bacterium, Smithella sp., Acidaminococcus sp., Lachnospiraceae bacterium, Candidatus Methanoplasma termitum, Eubacterium eligens, Moraxella bovoculi, Leptospira inadai, Porphyromonas crevioricanis, Prevotella disiens and Porphyromonas macacae, Succinivibrio dextrinosolvens, Prevotella disiens, Flavobacterium branchiophilum, Helcococcus kunzii, Eubacterium sp., Microgenomates (Roizmanbacteria) bacterium, Flavobacterium sp., Prevotella brevis, Moraxella caprae, Bacteroidetes oral, Porphyromonas cansulci, Synergistes jonesii, Prevotella bryantii, Anaerovibrio sp., Butyrivibrio fibrisolvens, Candidatus Methanomethylophilus, Butyrivibrio sp., Oribacterium sp., Pseudobutyrivibrio ruminis and Proteocatella sphenisci.

21. The method of claim 20, wherein said Cpf1 nickase protein is a FnCpf1 nickase and recognizes a PAM sequence of TTN, wherein N is A/C/G or T, or said Cpf1 nickase protein is a PaCpf1p, LbCpf1 or AsCpf1 nickase and recognizes a PAM sequence of TTTV, wherein V is A/C or G.

22. The method of claim 20, wherein said Cpf1 nickase protein has been modified and recognizes an altered PAM sequence.

23. The method of any of the preceding claims, wherein said target locus of interest is within a cell.

24. The method of claim 23, wherein said cell is a eukaryotic cell.

25. The method of claim 23, wherein said cell is a non-human animal cell, a human cell, or a plant cell.

26. The method of any of the preceding claims, wherein said target locus of interest is within an animal, within a plant, or comprised in a DNA molecule *in vitro*.

27. The method of any of the preceding claims, wherein said components (a), (b) and (c) are delivered to said cell as a ribonucleoprotein complex.

28. The method of any of the preceding claims, wherein said components (a), (b) and (c) are delivered to said cell as one or more polynucleotide molecules.

29. The method of claim 28, wherein said one or more polynucleotide molecules comprise one or more mRNA molecules encoding components (a) and/or (c).

30. The method of claim 28, wherein said one or more polynucleotide molecules are comprised within one or more vectors.

31. The method of claim 30, wherein said one or more polynucleotide molecules comprise one or more regulatory elements operably configured to express said Cpf1 nickase protein, said guide molecule, and said adenosine deaminase protein or catalytic domain thereof, optionally wherein said one or more regulatory elements comprise inducible promoters.

32. The method of any of claims 27-31, wherein said one or more polynucleotide molecules or said ribonucleoprotein complex are delivered via particles, vesicles, or one or more viral vectors.

33. The method of claim 32, wherein said particles comprise a lipid, a sugar, a metal or a protein.

34. The method of claim 33, wherein said particles comprise lipid nanoparticles.

35. The method of claim 32, wherein said vesicles comprise exosomes or liposomes.

36. The method of claim 32, wherein said one or more viral vectors comprise one or more of adenovirus, one or more lentivirus or one or more adeno-associated virus.

37. The method of any of the preceding claims, where said method modifies a cell, a cell line or an organism by manipulation of one or more target sequences at genomic loci of interest.

38. The method of claim 37, wherein said deamination of said Adenine at said target locus of interest remedies a disease caused by a G→A or C→T point mutation or a pathogenic SNP.

39. The method of claim 38, wherein said disease is selected from cancer, haemophilia, beta-thalassemia, Marfan syndrome and Wiskott-Aldrich syndrome.

40. The method of claim 37, wherein said deamination of said Adenine at said target locus of interest inactivates a target gene at said target locus.

41. A modified cell obtained from the method of any of the preceding claims, or progeny of said modified cell, wherein said cell comprises a hypoxanthine or a guanine in replace of said Adenine in said target locus of interest compared to a corresponding cell not subjected to the method.

42. The modified cell or progeny thereof of claim 41, wherein said cell is a eukaryotic cell.

43. The modified cell or progeny thereof of claim 41, wherein said cell is a non-human animal cell, a human cell, or a plant cell.

44. The modified cell or progeny thereof of claim 41, wherein said cell is a therapeutic T cell or antibody-producing B cell.

45. A non-human animal or a plant comprising said modified cell of claim 43.

46. A method for cell therapy, comprising administering to a patient in need thereof said modified cell of any of claims 41-44, wherein presence of said modified cell remedies a disease in the patient.

47. An engineered, non-naturally occurring system suitable for modifying an Adenine in a target locus of interest, comprising

a) a guide molecule which comprises a guide sequence linked to a direct repeat sequence, or a nucleotide sequence encoding said guide molecule;

b) a Cpf1 nickase protein, or a nucleotide sequence encoding said Cpf1 nickase protein;

c) an adenosine deaminase protein or catalytic domain thereof, or a nucleotide sequence encoding said adenosine deaminase protein or catalytic domain thereof;

wherein said adenosine deaminase protein or catalytic domain thereof is covalently or non-covalently linked to said Cpf1 nickase protein or said guide molecule or is adapted to link thereto after delivery;

wherein said guide sequence is capable of hybridizing with a target sequence comprising an Adenine on a first DNA strand at said target locus to form a heteroduplex, wherein said guide sequence comprises a non-pairing Cytosine at a position corresponding to said Adenine resulting in an A-C mismatch in the heteroduplex formed; and

wherein said Cpf1 nickase protein is capable of nicking a second DNA strand complementary to said first DNA strand.

48. An engineered, non-naturally occurring vector system suitable for modifying an Adenine in a target locus of interest, comprising the nucleotide sequences of a), b) and c) of claim 47.

49. The engineered, non-naturally occurring vector system of claim 48, comprising one or more vectors comprising:

a) a first regulatory element operably linked to a nucleotide sequence encoding said guide molecule which comprises said guide sequence,

b) a second regulatory element operably linked to a nucleotide sequence encoding said Cpf1 nickase protein; and

c) a nucleotide sequence encoding an adenosine deaminase protein or catalytic domain thereof which is under control of said first or second regulatory element or operably linked to a third regulatory element;

wherein, if said nucleotide sequence encoding an adenosine deaminase protein or catalytic domain thereof is operably linked to a third regulatory element, said adenosine deaminase protein or catalytic domain thereof is adapted to link to said guide molecule or said Cpf1 nickase protein after expression;

wherein components (a), (b) and (c) are located on the same or different vectors of the system.

50. An *in vitro* or *ex vivo* host cell or progeny thereof or cell line or progeny thereof comprising the system of any of claims 47-49, wherein optionally said cell is a eukaryotic cell.

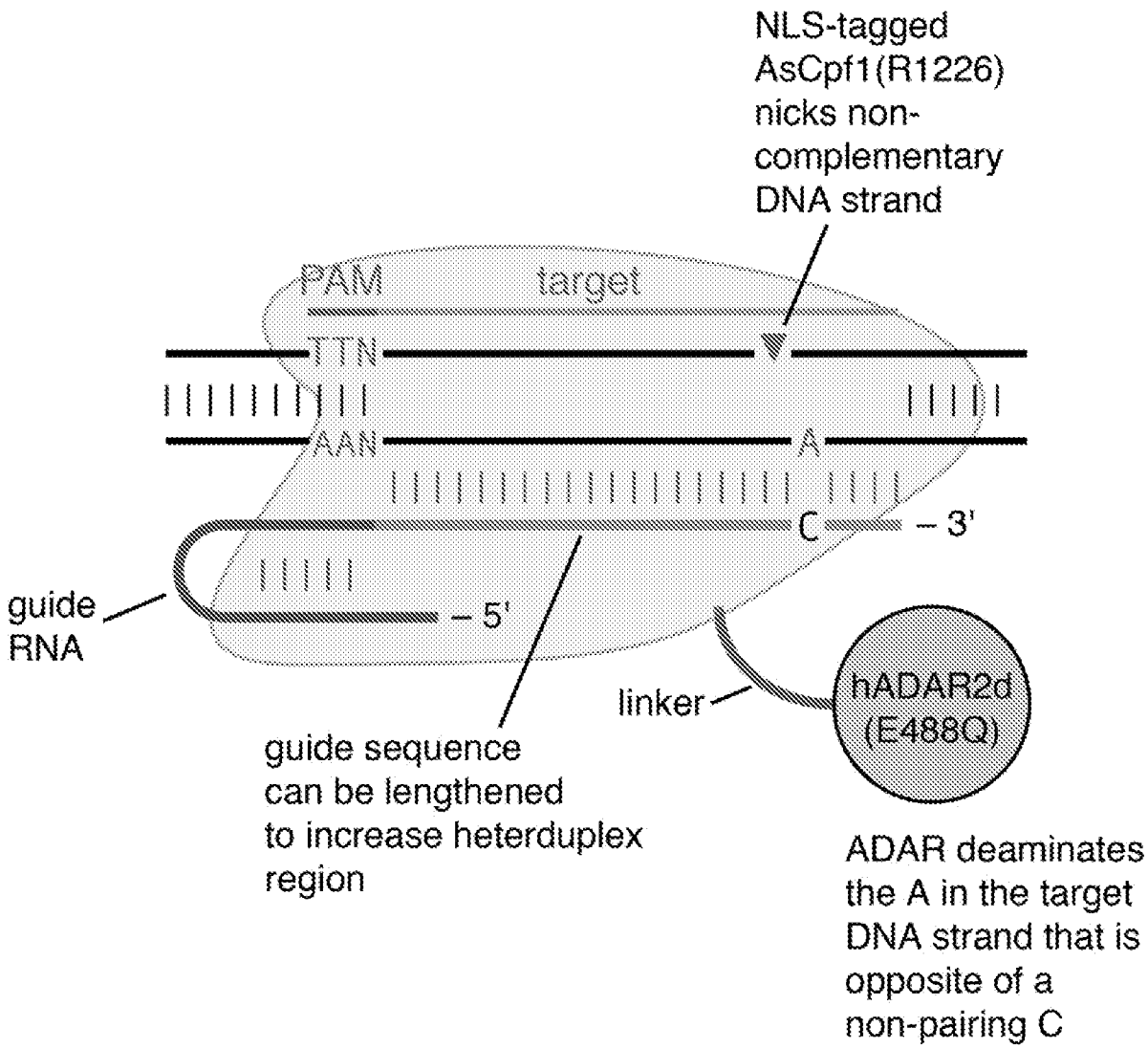

NLS-tagged
AsCpf1(R1226)
nicks non-
complementary
DNA strand
PAM
target
TTN
AAN
A
C
– 3'
– 5'
guide
RNA
linker
hADAR2d
(E488Q)
guide sequence
can be lengthened
to increase heterduplex
region
ADAR deaminates
the A in the target
DNA strand that is
opposite of a
non-pairing C

FIGURE 1

>NLS-FLAG-AsCpf1-linker-hADAR2d(wt)

MGPKKKRKVAAADYKDDDDKSMTQFEGFTNLYQVSKTLRFELIPQGKTLKHIQEQ
GFIEEDKARNDHYKELKPIIDRIYKTYADQCLQLVQLDWENLSAAIDSYRKEKTEET
RNALIEEQATYRNAIHDYFIGRTDNLTDAINKRHAEIYKGLFKAELFNGKVLKQLGT
VTTTEHENALLRSFDKFTTYFSGFYENRKNVFSAEDISTAIPHRIVQDNFPKFKENCH
IFTRLITAVPSLREHFENVKKAIGIFVSTSIEEVFSFPFYNQLLTQTQIDLYNQLLGGIS
REAGTEKIKGLNEVLNLAIQKNDETAHIIASLPHRFIPLFKQILSDRNTLSFILEEFKSD
EEVIQSFCKYKTLLRNENVLETAEALFNELNSIDLTHIFISHKKLETISSALCDHWDT
LRNALYERRISELTGKITKSAKEKVQRSLKHEDINLQEIISAAGKELSEAFKQKTSEIL
SHAHAALDQPLPTTLKKQEEKEILKSQLDSLLGLYHLLDWFAVDESNEVDPEFSAR
LTGIKLEMEPSLSFYNKARNYATKKPYSVEKFKLNFQMPTLASGWDVNKEKNNGA
ILFVKNGLYYLGIMPKQKGRYKALSFEPTEKTSEGFDKMYYDYFPDAAKMIPKCST
QLKAVTAHFQTHTTPILLSNNFIEPLEITKEIYDLNNPEKEPKKFQTAYAKKTGDQK
GYREALCKWIDFTRDFLSKYTKTTSIDLSSLRPSSQYKDLGEYYAELNPLLYHISFQR
IAEKEIMDAVETGKLYLFQIYNKDFAKGHHGKPNLHTLYWTGLFSPENLAKTSIKL
NGQAELFYRPKSRMKRMAHRLGEKMLNKKLKDQKTPIPDTLYQELYDYVNHRLS
HDLSDEARALLPNVITKEVSHEIIKDRRFTSDKFFFHVPITLNYQAANSPSKFNQRVN
AYLKEHPETPIIGIDRGERNLIYITVIDSTGKILEQRSLNTIQQFDYQKKLDNREKERV
AARQAWSVVGTIKDLKQGYLSQVIHEIVDLMIHYQAVVVLENLNFGFKSKRTGIAE
KAVYQQFEKMLIDKLNCLVLKDYPAEKVGGVLNPYQLTDQFTSFAKMGTQSGFLF
YVPAPYTSKIDPLTGFVDPFVWKTIKNHESRKHFLEGFDFLHYDVKTGDFILHFKM
NRNLSFQRGLPGFMPAWDIVFEKNETQFDAKGTPFIAGKRIVPVIENHRFTGRYRDL
YPANELIALLEEKGIVFRDGSNILPKLLENDDSHAIDTMVALIRSVLQMRNSNAATG
EDYINSPVRDLNGVCFDSRFQNPEWPMDADANGAYHIALKGQLLLNHLKESKDLK
LQNGISNQDWLAYIQELRNLEPGEKPYKCPECGKSFSQSGALTRHQRTHTRMDIED
EENMSSSSTDVKENRNLDNVSPKDGSTPGPGEGSQLSNGGGGGPGRKRPLEEGSNG
HSKYRLKKRRKTPGPVLPKNALMQLNEIKPGLQYTLLSQTGPVHAPLFVMSVEVN
GQVFEGSGPTKKKAKLHAAEKALRSFVQFPNASEAHLAMGRTLSVNTDFTSDQAD
FPDTLFNGFETPDKAEPPFYVGSNGDDSFSSSGDLSLSASPVPASLAQPPLPVLPPFPP
PSGKNPVMILNELRPGLKYDFLSESGESHAKSFVMSVVVDGQFFEGSGRNKKLAKA
RAAQSALAAIFNLHLDQTPSRQPIPSEGLQLHLPQVLADAVSRLVLGKFGDLTDNFS
SPHARRKVLAGVVMTTGTDVKDAKVISVSTGTKCINGEYMSDRGLALNDCHAEIIS
RRSLLRFLYTQLELYLNNKDDQKRSIFQKSERGGFRLKENVQFHLYISTSPCGDARIF
SPHEPILEEPADRHPNRKARGQLRTKIESGEGTIPVRSNASIQTWDGVLQGERLLTM
SCSDKIARWNVVGIQGSLLSIFVEPIYFSSIILGSLYHGDHLSRAMYQRISNIEDLPPL
YTLNKPLLSGISNAEARQPGKAPNFSVNWTVGDSAIEVINATTGKDELGRASRLCK
HALYCRWMRVHGKVPSHLLRSKITKPNVYHESKLAAKEYQAAKVH (SEQ ID
NO:79)

FIGURE 2

>NLS-FLAG-AsCpf1(R1226A)-linker-hADAR2d(E488Q)

MGPKKKRKVAAADYKDDDDKSMTQFEGFTNLYQVSKTLRFELIPQGKTLKHIQEQ
GFIEEDKARNDHYKELKPIIDRIYKTYADQCLQLVQLDWENLSAAIDSYRKEKTEET
RNALIEEQATYRNAIHDYFIGRTDNLTDAINKRHAEIYKGLFKAELFNGKVLKQLGT
VTTTEHENALLRSFDKFTTYFSGFYENRKNVFSAEDISTAIPHRIVQDNFPKFKENCH
IFTRLITAVPSLREHFENVKKAIGIFVSTSIEEVFSFPFYNQLLTQTQIDLYNQLLGGIS
REAGTEKIKGLNEVLNLAIQKNDETAHIIASLPHRFIPLFKQILSDRNTLSFILEEFKSD
EEVIQSFCKYKTLLRNENVLETAEALFNELNSIDLTHIFISHKKLETISSALCDHWDT
LRNALYERRISELTGKITKSAKEKVQRSLKHEDINLQEIISAAGKELSEAFKQKTSEIL
SHAHAALDQPLPTTLKKQEEKEILKSQLDSLLGLYHLLDWFAVDESNEVDPEFSAR
LTGIKLEMEPSLSFYNKARNYATKKPYSVEKFKLNFQMPTLASGWDVNKEKNNGA
ILFVKNGLYYLGIMPKQKGRYKALSFEPTEKTSEGFDKMYYDYFPDAAKMIPKCST
QLKAVTAHFQTHTTPILLSNNFIEPLEITKEIYDLNNPEKEPKKFQTAYAKKTGDQK
GYREALCKWIDFTRDFLSKYTKTTSIDLSSLRPSSQYKDLGEYYAELNPLLYHISFQR
IAEKEIMDAVETGKLYLFQIYNKDFAKGHHGKPNLHTLYWTGLFSPENLAKTSIKL
NGQAELFYRPKSRMKRMAHRLGEKMLNKKLKDQKTPIPDTLYQELYDYVNHRLS
HDLSDEARALLPNVITKEVSHEIIKDRRFTSDKFFFHVPITLNYQAANSPSKFNQRVN
AYLKEHPETPIIGIDRGERNLIYITVIDSTGKILEQRSLNTIQQFDYQKKLDNREKERV
AARQAWSVVGTIKDLKQGYLSQVIHEIVDLMIHYQAVVVLENLNFGFKSKRTGIAE
KAVYQQFEKMLIDKLNCLVLKDYPAEKVGGVLNPYQLTDQFTSFAKMGTQSGFLF
YVPAPYTSKIDPLTGFVDPFVWKTIKNHESRKHFLEGFDFLHYDVKTGDFILHFKM
NRNLSFQRGLPGFMPAWDIVFEKNETQFDAKGTPFIAGKRIVPVIENHRFTGRYRDL
YPANELIALLEEKGIVFRDGSNILPKLLENDDSHAIDTMVALIRSVLQMaNSNAATG
EDYINSPVRDLNGVCFDSRFQNPEWPMDADANGAYHIALKGQLLLNHLKESKDLK
LQNGISNQDWLAYIQELRNLEPGEKPYKCPECGKSFSQSGALTRHQRTHTRMDIED
EENMSSSSTDVKENRNLDNVSPKDGSTPGPGEGSQLSNGGGGGPGRKRPLEEGSNG
HSKYRLKKRRKTPGPVLPKNALMQLNEIKPGLQYTLLSQTGPVHAPLFVMSVEVN
GQVFEGSGPTKKKAKLHAAEKALRSFVQFPNASEAHLAMGRTLSVNTDFTSDQAD
FPDTLFNGFETPDKAEPPFYVGSNGDDSFSSSGDLSLSASPVPASLAQPPLPVLPPFPP
PSGKNPVMILNELRPGLKYDFLSESGESHAKSFVMSVVVDGQFFEGSGRNKKLAKA
RAAQSALAAIFNLHLDQTPSRQPIPSEGLQLHLPQVLADAVSRLVLGKFGDLTDNFS
SPHARRKVLAGVVMTTGTDVKDAKVISVSTGTKCINGEYMSDRGLALNDCHAEIIS
RRSLLRFLYTQLELYLNNKDDQKRSIFQKSERGGFRLKENVQFHLYISTSPCGDARIF
SPHEPILEEPADRHPNRKARGQLRTKIESGqGTIPVRSNASIQTWDGVLQGERLLTMS
CSDKIARWNVVGIQGSLLSIFVEPIYFSSIILGSLYHGDHLSRAMYQRISNIEDLPPLY
TLNKPLLSGISNAEARQPGKAPNFSVNWTVGDSAIEVINATTGKDELGRASRLCKH
ALYCRWMRVHGKVPSHLLRSKITKPNVYHESKLAAKEYQAAKVH (SEQ ID
NO:80)

FIGURE 3

>NLS-FLAG-AsCpf1(D908A)-linker-hADAR2d(E488Q)

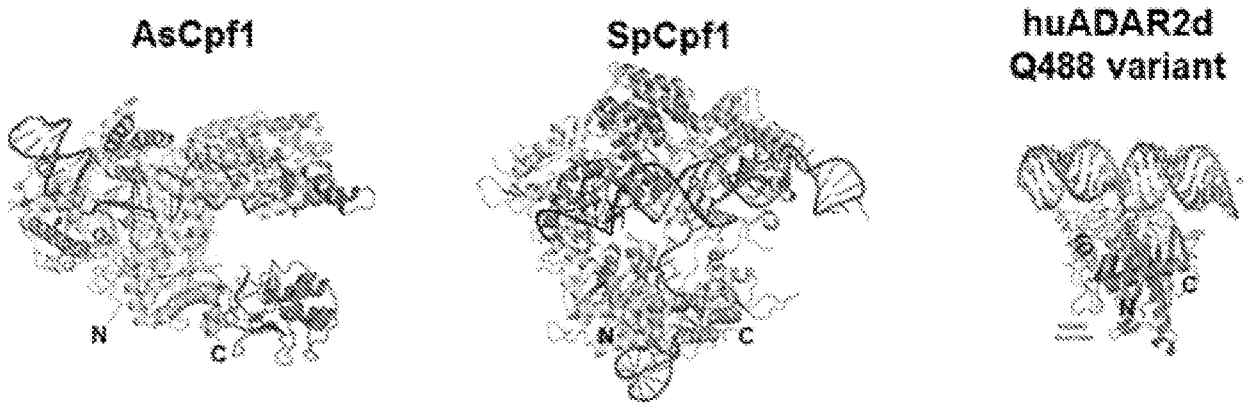
MGPKKKRKVAAADYKDDDDKSMTQFEGFTNLYQVSKTLRFELIPQGKTLKHIQEQ
GFIEEDKARNDHYKELKPIIDRIYKTYADQCLQLVQLDWENLSAAIDSYRKEKTEET
RNALIEEQATYRNAIHDYFIGRTDNLTDAINKRHAEIYKGLFKAELFNGKVLKQLGT
VTTTEHENALLRSFDKFTTYFSGFYENRKNVFSAEDISTAIPHRIVQDNFPKFKENCH
IFTRLITAVPSLREHFENVKKAIGIFVSTSIEEVFSFPFYNQLLTQTQIDLYNQLLGGIS
REAGTEKIKGLNEVLNLAIQKNDETAHIIASLPHRFIPLFKQILSDRNTLSFILEEFKSD
EEVIQSFCKYKTLLRNENVLETAEALFNELNSIDLTHIFISHKKLETISSALCDHWDT
LRNALYERRISELTGKITKSAKEKVQRSLKHEDINLQEIISAAGKELSEAFKQKTSEIL
SHAHAALDQPLPTTLKKQEEKEILKSQLDSLLGLYHLLDWFAVDESNEVDPEFSAR
LTGIKLEMEPSLSFYNKARNYATKKPYSVEKFKLNFQMPTLASGWDVNKEKNNGA
ILFVKNGLYYLGIMPKQKGRYKALSFEPTEKTSEGFDKMYYDYFPDAAKMIPKCST
QLKAVTAHFQTHTTPILLSNNFIEPLEITKEIYDLNNPEKEPKKFQTAYAKKTGDQK
GYREALCKWIDFTRDFLSKYTKTTSIDLSSLRPSSQYKDLGEYYAELNPLLYHISFQR
IAEKEIMDAVETGKLYLFQIYNKDFAKGHHGKPNLHTLYWTGLFSPENLAKTSIKL
NGQAELFYRPKSRMKRMAHRLGEKMLNKKLKDQKTPIPDTLYQELYDYVNHRLS
HDLSDEARALLPNVITKEVSHEIIKDRRFTSDKFFFHVPITLNYQAANSPSKFNQRVN
AYLKEHPETPIIGIaRGERNLIYITVIDSTGKILEQRSLNTIQQFDYQKKLDNREKERV
AARQAWSVVGTIKDLKQGYLSQVIHEIVDLMIHYQAVVVLENLNFGFKSKRTGIAE
KAVYQQFEKMLIDKLNCLVLKDYPAEKVGGVLNPYQLTDQFTSFAKMGTQSGFLF
YVPAPYTSKIDPLTGFVDPFVWKTIKNHESRKHFLEGFDFLHYDVKTGDFILHFKM
NRNLSFQRGLPGFMPAWDIVFEKNETQFDAKGTPFIAGKRIVPVIENHRFTGRYRDL
YPANELIALLEEKGIVFRDGSNILPKLLENDDSHAIDTMVALIRSVLQMRNSNAATG
EDYINSPVRDLNGVCFDSRFQNPEWPMDADANGAYHIALKGQLLLNHLKESKDLK
LQNGISNQDWLAYIQELRNLEPGEKPYKCPECGKSFSQSGALTRHQRTHTRMDIED
EENMSSSSTDVKENRNLDNVSPKDGSTPGPGEGSQLSNGGGGGPGRKRPLEEGSNG
HSKYRLKKRRKTPGPVLPKNALMQLNEIKPGLQYTLLSQTGPVHAPLFVMSVEVN
GQVFEGSGPTKKKAKLHAAEKALRSFVQFPNASEAHLAMGRTLSVNTDFTSDQAD
FPDTLFNGFETPDKAEPPFYVGSNGDDSFSSSGDLSLSASPVPASLAQPPLPVLPPFPP
PSGKNPVMILNELRPGLKYDFLSESGESHAKSFVMSVVVDGQFFEGSGRNKKLAKA
RAAQSALAAIFNLHLDQTPSRQPIPSEGLQLHLPQVLADAVSRLVLGKFGDLTDNFS
SPHARRKVLAGVVMTTGTDVKDAKVISVSTGTKCINGEYMSDRGLALNDCHAEIIS
RRSLLRFLYTQLELYLNNKDDQKRSIFQKSERGGFRLKENVQFHLYISTSPCGDARIF
SPHEPILEEPADRHPNRKARGQLRTKIESGqGTIPVRSNASIQTWDGVLQGERLLTMS
CSDKIARWNVVGIQGSLLSIFVEPIYFSSIILGSLYHGDHLSRAMYQRISNIEDLPPLY
TLNKPLLSGISNAEARQPGKAPNFSVNWTVGDSAIEVINATTGKDELGRASRLCKH
ALYCRWMRVHGKVPSHLLRSKITKPNVYHESKLAAKEYQAAKVH (SEQ ID
NO:81)

FIGURE 4

>NLS-FLAG-AsCpf1(E993A)-linker-hADAR2d(E488Q)

MGPKKKRKVAAADYKDDDDKSMTQFEGFTNLYQVSKTLRFELIPQGKTLKHIQEQ
GFIEEDKARNDHYKELKPIIDRIYKTYADQCLQLVQLDWENLSAAIDSYRKEKTEET
RNALIEEQATYRNAIHDYFIGRTDNLTDAINKRHAEIYKGLFKAELFNGKVLKQLGT
VTTTEHENALLRSFDKFTTYFSGFYENRKNVFSAEDISTAIPHRIVQDNFPKFKENCH
IFTRLITAVPSLREHFENVKKAIGIFVSTSIEEVFSFPFYNQLLTQTQIDLYNQLLGGIS
REAGTEKIKGLNEVLNLAIQKNDETAHIIASLPHRFIPLFKQILSDRNTLSFILEEFKSD
EEVIQSFCKYKTLLRNENVLETAEALFNELNSIDLTHIFISHKKLETISSALCDHWDT
LRNALYERRISELTGKITKSAKEKVQRSLKHEDINLQEIISAAGKELSEAFKQKTSEIL
SHAHAALDQPLPTTLKKQEEKEILKSQLDSLLGLYHLLDWFAVDESNEVDPEFSAR
LTGIKLEMEPSLSFYNKARNYATKKPYSVEKFKLNFQMPTLASGWDVNKEKNNGA
ILFVKNGLYYLGIMPKQKGRYKALSFEPTEKTSEGFDKMYYDYFPDAAKMIPKCST
QLKAVTAHFQTHTTPILLSNNFIEPLEITKEIYDLNNPEKEPKKFQTAYAKKTGDQK
GYREALCKWIDFTRDFLSKYTKTTSIDLSSLRPSSQYKDLGEYYAELNPLLYHISFQR
IAEKEIMDAVETGKLYLFQIYNKDFAKGHHGKPNLHTLYWTGLFSPENLAKTSIKL
NGQAELFYRPKSRMKRMAHRLGEKMLNKKLKDQKTPIPDTLYQELYDYVNHRLS
HDLSDEARALLPNVITKEVSHEIIKDRRFTSDKFFFHVPITLNYQAANSPSKFNQRVN
AYLKEHPETPIIGIDRGERNLIYITVIDSTGKILEQRSLNTIQQFDYQKKLDNREKERV
AARQAWSVVGTIKDLKQGYLSQVIHEIVDLMIHYQAVVVLaNLNFGFKSKRTGIAE
KAVYQQFEKMLIDKLNCLVLKDYPAEKVGGVLNPYQLTDQFTSFAKMGTQSGFLF
YVPAPYTSKIDPLTGFVDPFVWKTIKNHESRKHFLEGFDFLHYDVKTGDFILHFKM
NRNLSFQRGLPGFMPAWDIVFEKNETQFDAKGTPFIAGKRIVPVIENHRFTGRYRDL
YPANELIALLEEKGIVFRDGSNILPKLLENDDSHAIDTMVALIRSVLQMRNSNAATG
EDYINSPVRDLNGVCFDSRFQNPEWPMDADANGAYHIALKGQLLLNHLKESKDLK
LQNGISNQDWLAYIQELRNLEPGEKPYKCPECGKSFSQSGALTRHQRTHTRMDIED
EENMSSSSTDVKENRNLDNVSPKDGSTPGPGEGSQLSNGGGGGPGRKRPLEEGSNG
HSKYRLKKRRKTPGPVLPKNALMQLNEIKPGLQYTLLSQTGPVHAPLFVMSVEVN
GQVFEGSGPTKKKAKLHAAEKALRSFVQFPNASEAHLAMGRTLSVNTDFTSDQAD
FPDTLFNGFETPDKAEPPFYVGSNGDDSFSSSGDLSLSASPVPASLAQPPLPVLPPFPP
PSGKNPVMILNELRPGLKYDFLSESGESHAKSFVMSVVVDGQFFEGSGRNKKLAKA
RAAQSALAAIFNLHLDQTPSRQPIPSEGLQLHLPQVLADAVSRLVLGKFGDLTDNFS
SPHARRKVLAGVVMTTGTDVKDAKVISVSTGTKCINGEYMSDRGLALNDCHAEIIS
RRSLLRFLYTQLELYLNNKDDQKRSIFQKSERGGFRLKENVQFHLYISTSPCGDARIF
SPHEPILEEPADRHPNRKARGQLRTKIESGqGTIPVRSNASIQTWDGVLQGERLLTMS
CSDKIARWNVVGIQGSLLSIFVEPIYFSSIILGSLYHGDHLSRAMYQRISNIEDLPPLY
TLNKPLLSGISNAEARQPGKAPNFSVNWTVGDSAIEVINATTGKDELGRASRLCKH
ALYCRWMRVHGKVPSHLLRSKITKPNVYHESKLAAKEYQAAKVH (SEQ ID
NO:82)

FIGURE 5



N-term ADAR: huADAR2d-NLS-linker-NLS-Cas9/Cpf1-NLS

C-term ADAR: NLS-Cas9/Cpf1-NLS-linker-huADAR2d-NLS

Linker 2: GGGGSGGGSGGGSGGGSGGGG (21aa) (SEQ ID NO:83)

Linker 3: SGSETPGTSESATPESGGSGGSGSETPGTSESATPES (37aa) (SEQ ID NO:84)

FIGURE 6

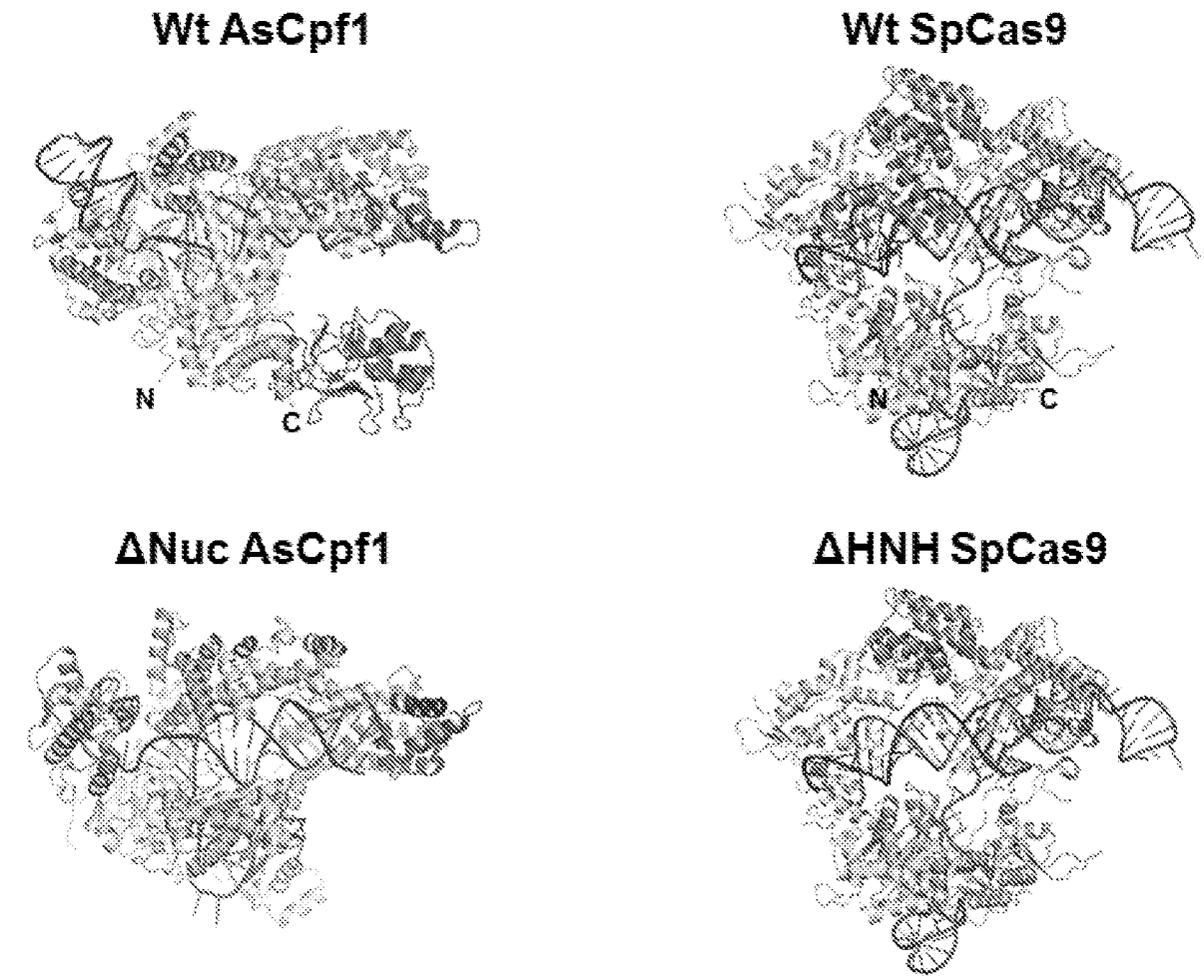
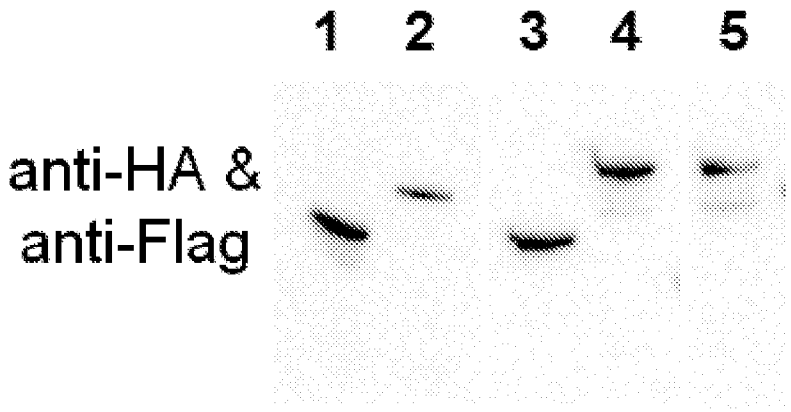
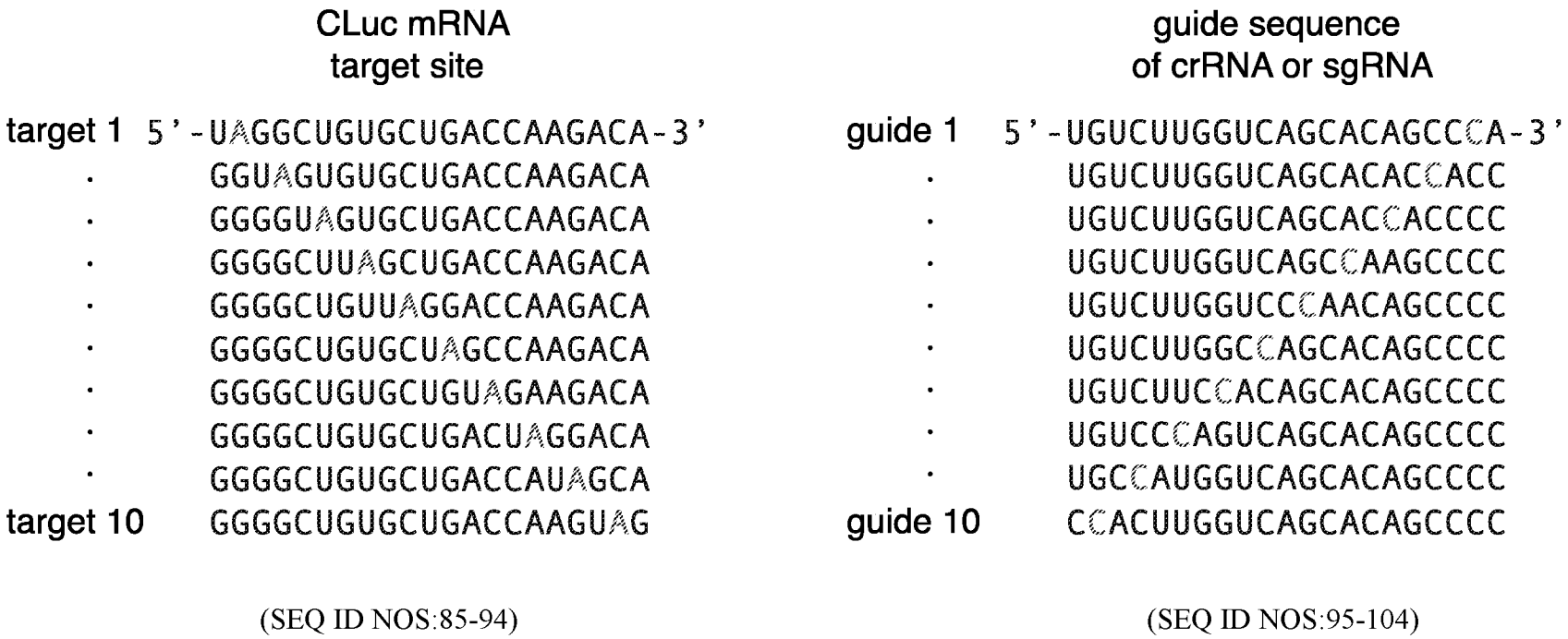

Wt AsCpf1
N
C
Wt SpCas9
N
C
ΔNuc AsCpf1
Amino acids 1076 to 1258
were replaced with a GSGG linker
ΔHNH SpCas9
Amino acids 769 to 918
were replaced with a GGSGGS linker

FIGURE 7



1 = delta HNH SpCas9
2 = delta HNH SpCAs9-link1-huADAR2d
3 = delta Nuc AsCpf1
4 = huADAR2d-link3-delta Nuc AsCpf1
5 = huADAR2d-link3-delta Nuc AsCpf1

FIGURE 8



(SEQ ID NOS:85-94)

(SEQ ID NOS:95-104)

FIGURE 9

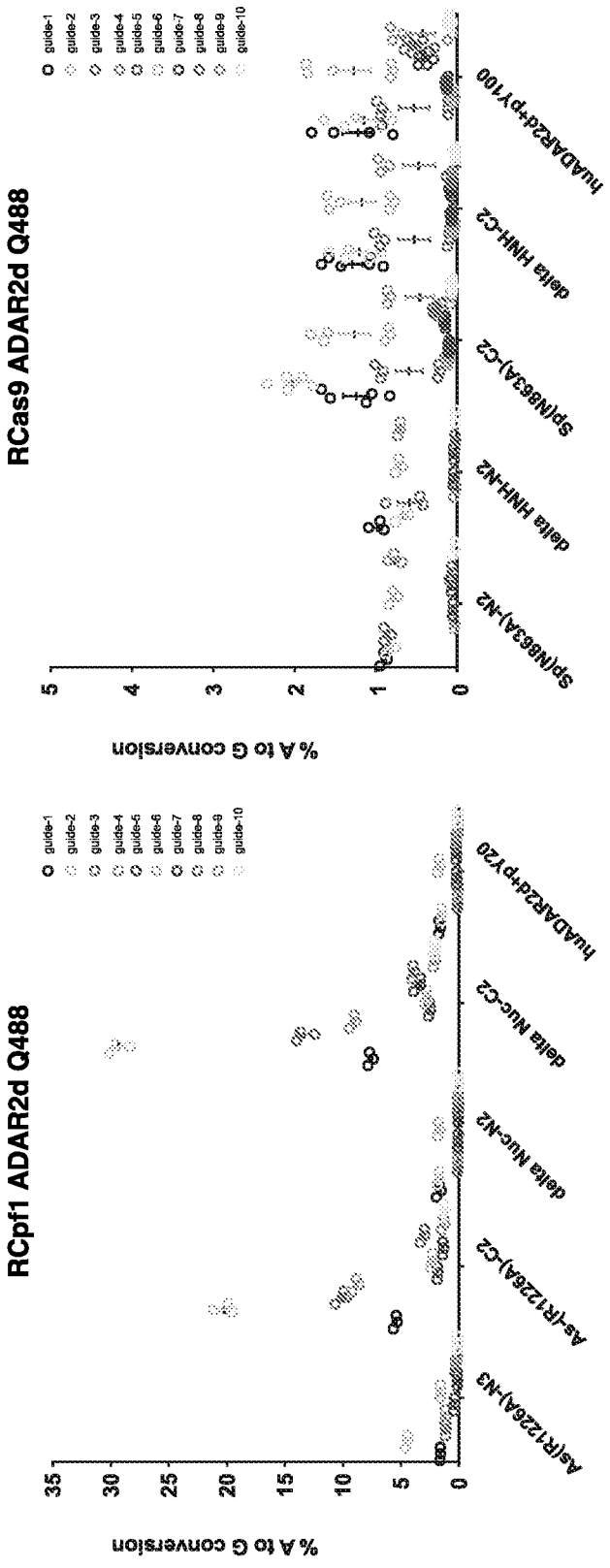

RCas9 ADAR2d Q488
% A to G conversion
guide-1
guide-2
guide-3
guide-4
guide-5
guide-6
guide-7
guide-8
guide-9
guide-10
SpCas9(N863A)-N2
delta HNH-N2
SpCas9(N863A)-C2
delta HNH-C2
huADAR2d-pY100
RCpf1 ADAR2d Q488
% A to G conversion
AsCas12a(R1226A)-N3
As-Cas12a(R1226A)-C2
delta Nuc-N2
delta Nuc-C2
huADAR2d-pY20

FIGURE 10

target site
(Human DNMT1
in HEK293 cells)

5'-..CTGATGGTCCATGTCTGTTAC..-3' (SEQ ID NO:105)
3'-..GACTACCAGGTACAGACAATG..-5' (SEQ ID NO:106)

targeted A

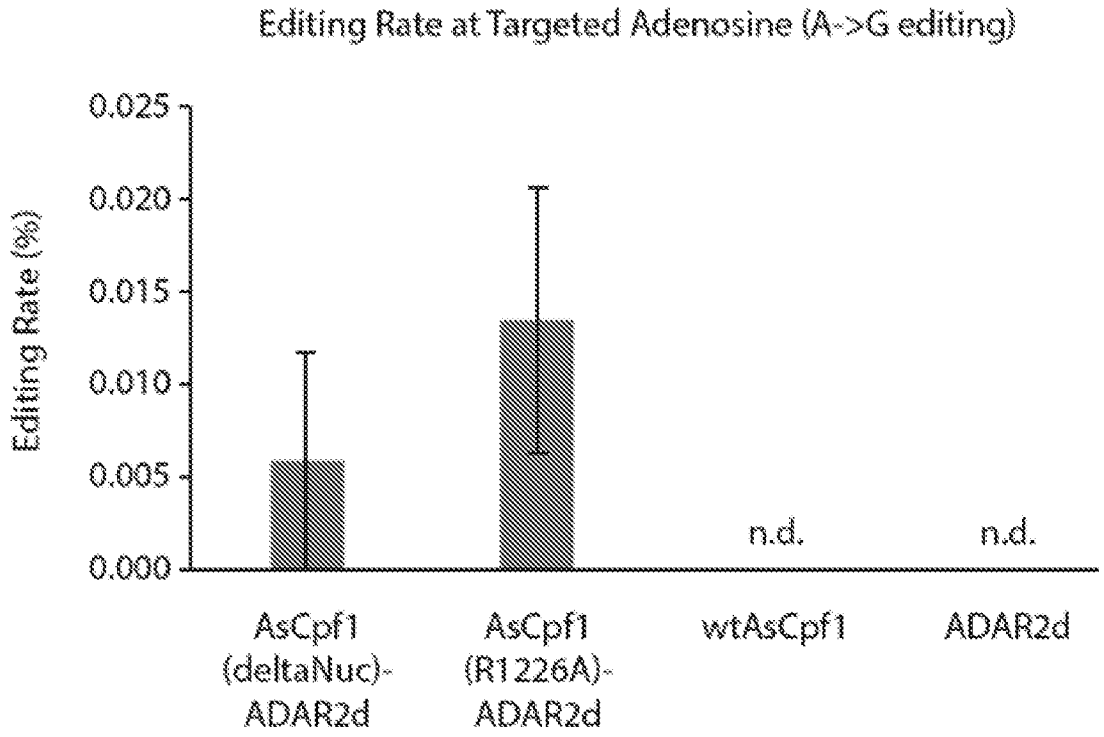


FIGURE 11

INTERNATIONAL SEARCH REPORT

International application No
PCT/US2018/033394

A. CLASSIFICATION OF SUBJECT MATTER
INV. C12N15/10 C12N9/22 C12N9/78 C12N15/62
ADD.

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)
C12N

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practicable, search terms used)
EPO-Internal, BIOSIS, EMBASE, WPI Data

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category*	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X Y	WO 2015/089406 A1 (HARVARD COLLEGE [US]) 18 June 2015 (2015-06-18) paragraphs [0046], [0050], [0114], [0123], [0124]; claims 1-3,9-16,22-62; sequences 35,36 -----	41-45 1-40, 46-50
Y	MALZAHN AIMEE ET AL: "Plant genome editing with TALEN and CRISPR", CELL BIOSCIENCE, vol. 7, 24 April 2017 (2017-04-24), XP002785201, ISSN: 2045-3701 page 12, column 1, paragraph 3 - page 12, column 2, paragraph 1 ----- -/--	1-40, 46-50

[X] Further documents are listed in the continuation of Box C.

[X] See patent family annex.

* Special categories of cited documents :

"A" document defining the general state of the art which is not considered to be of particular relevance

"E" earlier application or patent but published on or after the international filing date

"L" document which may throw doubts on priority claim(s) or which is cited to establish the publication date of another citation or other special reason (as specified)

"O" document referring to an oral disclosure, use, exhibition or other means

"P" document published prior to the international filing date but later than the priority date claimed

"T" later document published after the international filing date or priority date and not in conflict with the application but cited to understand the principle or theory underlying the invention

"X" document of particular relevance; the claimed invention cannot be considered novel or cannot be considered to involve an inventive step when the document is taken alone

"Y" document of particular relevance; the claimed invention cannot be considered to involve an inventive step when the document is combined with one or more other such documents, such combination being obvious to a person skilled in the art

"&" document member of the same patent family

Date of the actual completion of the international search	Date of mailing of the international search report
4 October 2018	24/10/2018

Name and mailing address of the ISA/	Authorized officer
European Patent Office, P.B. 5818 Patentlaan 2 NL - 2280 HV Rijswijk Tel. (+31-70) 340-2040, Fax: (+31-70) 340-3016	Seroz, Thierry

3

Form PCT/ISA/210 (second sheet) (April 2005)

C(Continuation). DOCUMENTS CONSIDERED TO BE RELEVANT

Category*	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
Y	DAESIK KIM ET AL: "Genome-wide target specificities of CRISPR RNA-guided programmable deaminases", NATURE BIOTECHNOLOGY, vol. 35, no. 5, 10 April 2017 (2017-04-10) , pages 475-480, XP055383071, ISSN: 1087-0156, DOI: 10.1038/nbt.3852 page 475, column 1, paragraph 2	5,6
A	US 2015/071903 A1 (LIU DAVID R [US] ET AL) 12 March 2015 (2015-03-12) paragraphs [0011], [0012], [0149]; claims 1-30	1-50
A	YUXUAN ZHENG ET AL: "DNA editing in DNA/RNA hybrids by adenosine deaminases that act on RNA", NUCLEIC ACIDS RESEARCH, vol. 45, no. 6, GKX050, 28 January 2017 (2017-01-28), pages 3369-3377, XP055404026, ISSN: 0305-1048, DOI: 10.1093/nar/gkx050 the whole document	1-50
X,P	MATSOUKAS IANIS G: "Commentary: Programmable base editing of A center dot T to G center dot C in genomic DNA without DNA cleavage", FRONTIERS IN GENETICS, vol. 9, 7 February 2018 (2018-02-07), XP002785202, the whole document	1-50
X,P	GAUDELLI NICOLE M ET AL: "Programmable base editing of A.T to G.C in genomic DNA without DNA cleavage (Includes Methods)", NATURE, vol. 551, no. 7681, 23 November 2017 (2017-11-23), pages 464-471+16PP, XP002785203, LONDON ISSN: 0028-0836, DOI: 10.1038/nature24644 the whole document	1-50

INTERNATIONAL SEARCH REPORT

Information on patent family members

International application No
PCT/US2018/033394

Patent document cited in search report	Publication date	Patent family member(s)	Publication date
WO 2015089406 A1	18-06-2015	AU 2014362208 A1	30-06-2016
		CA 2933625 A1	18-06-2015
		CN 105934516 A	07-09-2016
		EP 3080265 A1	19-10-2016
		JP 2017500035 A	05-01-2017
		US 2015165054 A1	18-06-2015
		US 2015166980 A1	18-06-2015
		US 2015166981 A1	18-06-2015
		US 2015166982 A1	18-06-2015
		US 2015166983 A1	18-06-2015
		US 2015166984 A1	18-06-2015
		US 2015166985 A1	18-06-2015
		US 2016304846 A1	20-10-2016
		WO 2015089406 A1	18-06-2015
US 2015071903 A1	12-03-2015	AU 2014315132 A1	24-03-2016
		AU 2015338968 A1	18-05-2017
		AU 2018208708 A1	16-08-2018
		CA 2923409 A1	12-03-2015
		CA 2965967 A1	06-05-2016
		CN 105745221 A	06-07-2016
		EP 3041858 A2	13-07-2016
		JP 2016534132 A	04-11-2016
		KR 20160050070 A	10-05-2016
		SG 11201601658Y A	28-04-2016
		US 2015071903 A1	12-03-2015
		US 2015071906 A1	12-03-2015
		US 2015118216 A1	30-04-2015
		US 2016200779 A1	14-07-2016
		US 2018236081 A1	23-08-2018
		WO 2015035136 A2	12-03-2015