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(54) Title: METHODS OF TREATING CLONAL HEMATOPOIESIS OF INDETERMINATE POTENTIAL (CHIP) WITH LYMPHOCYTE ANTIGEN 75 (LY75), CLUSTER OF DIFFERENTIATION 164 (CD164), OR POLY(ADP-RIBOSE) POLYMERASE 1 (PARP1) INHIBITORS

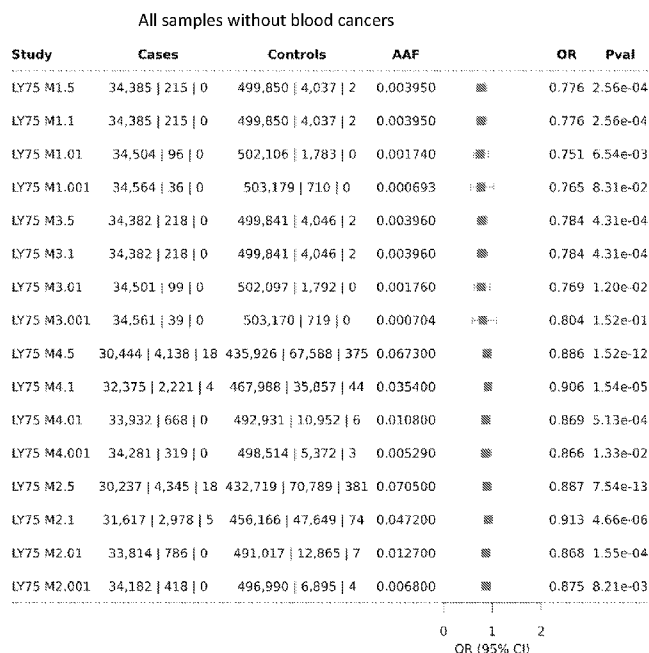


Figure 1

(57) Abstract: Methods of treating subjects having clonal hematopoiesis of indeterminate potential (CHIP) with lymphocyte antigen 75 (LY75), Cluster of Differentiation 164 (CD164), or Poly(ADP-Ribose) Polymerase 1 (PARP1) inhibitors, and methods of identifying subjects having an increased risk of developing CHIP are provided herein.

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**Methods Of Treating Clonal Hematopoiesis Of Indeterminate Potential (CHIP) With
Lymphocyte Antigen 75 (LY75), Cluster Of Differentiation 164 (CD164), Or Poly(ADP-Ribose)
Polymerase 1 (PARP1) Inhibitors**

5 Reference To Sequence Listing

This application includes a Sequence Listing filed electronically as an XML file named 381203582SEQ, created on September 11, 2022, with a size of 1,547 kilobytes. The Sequence Listing is incorporated herein by reference.

10 Field

The present disclosure relates generally to the treatment of subjects having clonal hematopoiesis of indeterminate potential (CHIP) with Lymphocyte Antigen 75 (LY75), Cluster of Differentiation 164 (CD164), or Poly(ADP-Ribose) Polymerase 1 (PARP1) inhibitors, and methods of identifying subjects having an increased risk of developing CHIP.

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Background

CHIP is a genetically defined phenotype reflecting age-related changes to hematopoietic stem cells (HSCs). As a person ages, their HSCs accumulate mutations as a result of DNA replication error and DNA damage repair (so called somatic mutations, such as those acquired after birth). Thus, prevalence rises with age and is roughly 10% among persons aged 70 to 80. Patients undergoing molecular genetic investigation for cytopenia (anemia, leukopenia, thrombocytopenia) are the most likely to be given this diagnosis. Some of these mutations confer growth advantages, which result in: increased proliferation of these cells relative to other cells, increase in frequency of these mutations, and accumulation of additional mutations that drive neoplastic changes. A subset of genes are strongly recurrently mutated along with clonal hematopoiesis; these are considered "CHIP genes" and they include: DNA Methyltransferase 3 Alpha (DNMT3A), Tet Methylcytosine Dioxygenase 2 (TET2), ASXL Transcriptional Regulator 1 (ASXL1), Janus kinase 2 (JAK2), and Splicing factor 3B subunit 1 (SF3B1). CpG => TpG mutations are very common in CHIP. In addition to the identification in blood DNA of specific recurrent mutations, the clinical definition of CHIP requires the absence of dysplasia and leukemia (< 20% blasts). CHIP is associated with increased risk of hematologic cancers, such as myeloid or lymphoid neoplasia, and with increased risk of atherosclerotic

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cardiovascular disease, such as coronary heart disease, myocardial infarction, and severe calcified aortic valve stenosis.

LY75 is an endocytic receptor that captures antigens from the extracellular space and directs them to a specialized antigen-processing compartment for antigen processing, presentation, and cross-presentation. LY75 can cause reduced proliferation of B-lymphocytes. LY75 is expressed on human dendritic cells, monocytes, B cells, T cells, NK cells, and thymic epithelial cells.

CD164 is an adhesive glycoprotein expressed by HSCs and bone marrow stromal cells that acts as a regulator of hematopoiesis. CD164 belongs to the sialomucin family of secreted or membrane-associated mucins that regulate proliferation, adhesion, and migration of HSCs.

PARP1 is a DNA repair protein involved in PAR polymerization, and recruits an array of repair molecules to support single and double strand repair, and chromatin remodeling in the context of NER. PARP1 modifies various nuclear proteins by poly(ADP-ribosyl)ation of glutamate, aspartate, serine, or tyrosine residues. The modification is dependent on DNA and is involved in the regulation of various important cellular processes such as differentiation, proliferation, and tumor transformation and also in the regulation of the molecular events involved in the recovery of cells from DNA damage.

Summary

The present disclosure provides methods of preventing or reducing the development of CHIP in a subject, the methods comprising administering an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject.

The present disclosure also provides methods of treating a subject with a therapeutic agent that prevents or reduces development of CHIP, wherein the subject has CHIP or is at risk of developing CHIP, the methods comprising the steps of: determining whether the subject has an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and/or a PARP1 variant nucleic acid molecule by: obtaining or having obtained a biological sample from the subject; and performing or having performed a sequence analysis on the biological sample to determine if the subject has a genotype comprising the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule; and administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount to a subject that is LY75 reference, CD164

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reference, and/or PARP1 reference, and/or administering an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject; administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is heterozygous for the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule, and/or administering an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject; or administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is homozygous for the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule; wherein the presence of a genotype having the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule indicates the subject has a decreased risk of developing CHIP.

The present disclosure also provides methods of identifying a subject having an increased risk of developing CHIP, the methods comprising: determining or having determined the presence or absence of an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and/or a PARP1 variant nucleic acid molecule in a biological sample obtained from the subject; wherein: when the subject is LY75 reference, CD164 reference, and/or PARP1 reference, then the subject has an increased risk of developing CHIP; and when the subject is heterozygous or homozygous for the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule, then the subject has a decreased risk of developing CHIP.

The present disclosure also provides therapeutic agents that prevent or reduce CHIP for use in the prevention or reduction of CHIP in a subject identified as having: an LY75 variant genomic nucleic acid molecule, a CD164 variant genomic nucleic acid molecule, and/or a PARP1 variant genomic nucleic acid molecule; an LY75 variant mRNA molecule, a CD164 variant mRNA molecule, and/or a PARP1 variant mRNA molecule; or an LY75 variant cDNA molecule, a CD164 variant cDNA molecule, and/or a PARP1 variant cDNA molecule.

The present disclosure also provides an LY75 inhibitor for use in the prevention or reduction of CHIP in a subject that: a) is reference for an LY75 genomic nucleic acid molecule, an LY75 mRNA molecule, or an LY75 cDNA molecule; or b) is heterozygous for: i) an LY75 variant

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genomic nucleic acid molecule; ii) an LY75 variant mRNA molecule; or iii) an LY75 variant cDNA molecule.

The present disclosure also provides a CD164 inhibitor for use in the prevention or reduction of CHIP in a subject that: a) is reference for a CD164 genomic nucleic acid molecule, a
5 CD164 mRNA molecule, or a CD164 cDNA molecule; or b) is heterozygous for: i) a CD164 variant genomic nucleic acid molecule; ii) a CD164 variant mRNA molecule; or iii) a CD164 variant cDNA molecule.

The present disclosure also provides a PARP1 inhibitor for use in the prevention or reduction of CHIP in a subject that: a) is reference for a PARP1 genomic nucleic acid molecule, a
10 PARP1 mRNA molecule, or a PARP1 cDNA molecule; or b) is heterozygous for: i) a PARP1 variant genomic nucleic acid molecule; ii) a PARP1 variant mRNA molecule; or iii) a PARP1 variant cDNA molecule.

The present disclosure also provides methods of identifying a subject at risk of developing lung cancer, the method comprising: determining or having determined the
15 presence or absence of one or more CHIP somatic mutations in DNA Methyltransferase 3 Alpha (DNMT3A) and/or ASXL Transcriptional Regulator 1 (ASXL1) in a biological sample obtained from the subject; wherein: when the subject has a CHIP somatic mutation in DNMT3A and/or ASXL1, the subject has an increased risk of developing lung cancer; and when the subject does not have a CHIP somatic mutation in DNMT3A and/or ASXL1, the subject does not have an
20 increased risk of developing lung cancer.

Brief Description Of The Drawings

The accompanying figures, which are incorporated in and constitute a part of this specification, illustrate several features of the present disclosure.

25 The patent or application file contains at least one drawing executed in color. Copies of this patent or patent application publication with color drawing(s) will be provided by the Office upon request and payment of the necessary fee.

Figure 1 shows LY75 burden masks analysis of CHIP risk.

Figure 2 shows the effect of the ly75 locus stratified by CHIP gene mutation carrier.

30 Figure 3 shows CHIP minus malignant blood LY75-CD302 masks.

Figure 4 shows that the PARP 1:226367601:A:C missense variant significantly associated with CHIP.

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Figure 5 shows PARP missense variants significantly associate with CHIP.

Figure 6 shows that CHIP carriers are at an elevated risk of developing solid tumors.

Figure 7 shows functional modeling of identified CHIP-GWAS locus; Panels A and B show systematic tracking of HDR and INDEL percentages in PARPi treated cells; cells were
5 treated with PARPi for 8 days and then subjected to Sanger sequencing across each locus to determine population dynamics; (Panel A) RPE-1 hTERT cells harboring CRISPR mediated knock-in of DNMT3A-R882H allele; (Panel B) CRISPR-mediated TET2-deficiency in HEK293T cells.

Figure 8 shows Forest plots reflecting the protective associations of two LY75 missense variants (rs78446341-A, rs147820690-T) with the DNMT3A-CHIP overall CHIP phenotype in the
10 UKB and GHS cohorts.

Description

Various terms relating to aspects of the present disclosure are used throughout the specification and claims. Such terms are to be given their ordinary meaning in the art, unless
15 otherwise indicated. Other specifically defined terms are to be construed in a manner consistent with the definitions provided herein.

Unless otherwise expressly stated, it is in no way intended that any method or aspect set forth herein be construed as requiring that its steps be performed in a specific order. Accordingly, where a method claim does not specifically state in the claims or descriptions that
20 the steps are to be limited to a specific order, it is in no way intended that an order be inferred, in any respect. This holds for any possible non-expressed basis for interpretation, including matters of logic with respect to arrangement of steps or operational flow, plain meaning derived from grammatical organization or punctuation, or the number or type of aspects described in the specification.

25 As used herein, the singular forms “a,” “an” and “the” include plural referents unless the context clearly dictates otherwise.

As used herein, the term “about” means that the recited numerical value is approximate and small variations would not significantly affect the practice of the disclosed embodiments. Where a numerical value is used, unless indicated otherwise by the context, the
30 term “about” means the numerical value can vary by $\pm 10\%$ and remain within the scope of the disclosed embodiments.

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As used herein, the term “comprising” may be replaced with “consisting” or “consisting essentially of” in particular embodiments as desired.

As used herein, the term “isolated”, in regard to a nucleic acid molecule or a polypeptide, means that the nucleic acid molecule or polypeptide is in a condition other than its native environment, such as apart from blood and/or animal tissue. In some embodiments, an isolated nucleic acid molecule or polypeptide is substantially free of other nucleic acid molecules or other polypeptides, particularly other nucleic acid molecules or polypeptides of animal origin. In some embodiments, the nucleic acid molecule or polypeptide can be in a highly purified form, i.e., greater than 95% pure or greater than 99% pure. When used in this context, the term “isolated” does not exclude the presence of the same nucleic acid molecule or polypeptide in alternative physical forms, such as dimers or Alternately phosphorylated or derivatized forms.

As used herein, the terms “nucleic acid”, “nucleic acid molecule”, “nucleic acid sequence”, “polynucleotide”, or “oligonucleotide” can comprise a polymeric form of nucleotides of any length, can comprise DNA and/or RNA, and can be single-stranded, double-stranded, or multiple stranded. One strand of a nucleic acid also refers to its complement.

As used herein, the term “subject” includes any animal, including mammals. Mammals include, but are not limited to, farm animals (such as, for example, horse, cow, pig), companion animals (such as, for example, dog, cat), laboratory animals (such as, for example, mouse, rat, rabbits), and non-human primates. In some embodiments, the subject is a human. In some embodiments, the human is a patient under the care of a physician.

It has been observed in accordance with the present disclosure that an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and a PARP1 variant nucleic acid molecule (whether these variations are homozygous or heterozygous in a particular subject) associate with a decreased risk of developing CHIP or CHIP-related disorders. The identification by the present disclosure of the association between additional variants and gene burden masks indicates that one or more of LY75, CD164, and PARP1 themselves (rather than linkage disequilibrium with variants in another gene) are responsible for a protective effect in CHI) and CHIP-related disorders.

Therefore, subjects that are LY75 reference, or heterozygous for an LY75 variant nucleic acid molecule, may be treated with an LY75 inhibitor; subjects that are CD164 reference, or heterozygous for a CD164 variant nucleic acid molecule, may be treated with a

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CD164 inhibitor; and subjects that are PARP1 reference, or heterozygous for a PARP1 variant nucleic acid molecules, may be treated with a PARP1 inhibitor, such that CHIP is prevented or inhibited and CHIP-related disorders are inhibited or prevented, the symptoms thereof are reduced or prevented, and/or development of symptoms is repressed or prevented. It is also
5 believed that such subjects having CHIP may further be treated with therapeutic agents that treat or inhibit CHIP or CHIP-related disorders.

For purposes of the present disclosure, any particular subject, such as a human, can be categorized as having one of three LY75 genotypes: i) LY75 reference; ii) heterozygous for an LY75 variant nucleic acid molecule; or iii) homozygous for an LY75 variant nucleic acid molecule.

10 A subject is LY75 reference when the subject does not have a copy of an LY75 variant nucleic acid molecule. A subject is heterozygous for an LY75 variant nucleic acid molecule when the subject has a single copy of an LY75 variant nucleic acid molecule.

In any of the embodiments described herein, an LY75 variant nucleic acid molecule is any nucleic acid molecule (such as, a genomic nucleic acid molecule, an mRNA molecule, or a
15 cDNA molecule) that is a missense variant, a splice-site variant, a stop-gain variant, a start-loss variant, a stop-loss variant, a frameshift variant, an in-frame indel variant, or a variant that encodes a truncated LY75 polypeptide. In any of the embodiments described herein, an LY75 variant nucleic acid molecule can also be any nucleic acid molecule (such as, a genomic nucleic acid molecule, an mRNA molecule, or a cDNA molecule) that encodes an LY75 polypeptide
20 having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function. A subject who has an LY75 polypeptide having a partial loss-of-function (or predicted partial loss-of-function) is hypomorphic for LY75. A subject is homozygous for an LY75 variant nucleic acid molecule when the subject has two copies (same or different) of an LY75 variant nucleic acid molecule.

25 For purposes of the present disclosure, any particular subject, such as a human, can be categorized as having one of three CD164 genotypes: i) CD164 reference; ii) heterozygous for a CD164 variant nucleic acid molecule; or iii) homozygous for a CD164 variant nucleic acid molecule. A subject is CD164 reference when the subject does not have a copy of a CD164 variant nucleic acid molecule. A subject is heterozygous for a CD164 variant nucleic acid
30 molecule when the subject has a single copy of a CD164 variant nucleic acid molecule.

In any of the embodiments described herein, a CD164 variant nucleic acid molecule is any nucleic acid molecule (such as, a genomic nucleic acid molecule, an mRNA molecule, or a

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cDNA molecule) that is a missense variant, a splice-site variant, a stop-gain variant, a start-loss variant, a stop-loss variant, a frameshift variant, an in-frame indel variant, or a variant that encodes a truncated CD164 polypeptide. In any of the embodiments described herein, a CD164 variant nucleic acid molecule can also be any nucleic acid molecule (such as, a genomic nucleic acid molecule, an mRNA molecule, or a cDNA molecule) that encodes a CD164 polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function. A subject who has a CD164 polypeptide having a partial loss-of-function (or predicted partial loss-of-function) is hypomorphic for CD164. A subject is homozygous for a CD164 variant nucleic acid molecule when the subject has two copies (same or different) of a CD164 variant nucleic acid molecule.

For purposes of the present disclosure, any particular subject, such as a human, can be categorized as having one of three PARP1 genotypes: i) PARP1 reference; ii) heterozygous for a PARP1 variant nucleic acid molecule; or iii) homozygous for a CD164 variant nucleic acid molecule. A subject is PARP1 reference when the subject does not have a copy of a PARP1 variant nucleic acid molecule. A subject is heterozygous for a PARP1 variant nucleic acid molecule when the subject has a single copy of a PARP1 variant nucleic acid molecule.

In any of the embodiments described herein, a PARP1 variant nucleic acid molecule is any nucleic acid molecule (such as, a genomic nucleic acid molecule, an mRNA molecule, or a cDNA molecule) that is a missense variant, a splice-site variant, a stop-gain variant, a start-loss variant, a stop-loss variant, a frameshift variant, an in-frame indel variant, or a variant that encodes a truncated PARP1 polypeptide. In any of the embodiments described herein, a PARP1 variant nucleic acid molecule can also be any nucleic acid molecule (such as, a genomic nucleic acid molecule, an mRNA molecule, or a cDNA molecule) that encodes a PARP1 polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function. A subject who has a PARP1 polypeptide having a partial loss-of-function (or predicted partial loss-of-function) is hypomorphic for PARP1. A subject is homozygous for a PARP1 variant nucleic acid molecule when the subject has two copies (same or different) of a PARP1 variant nucleic acid molecule.

For subjects that are genotyped or determined to be LY75, CD164, and PARP1 reference, such subjects have an increased risk of developing CHIP and CHIP-related disorders, such as a hematologic cancer, a myeloid neoplasia, a lymphoid neoplasia, an atherosclerotic cardiovascular disease, a coronary heart disease, a myocardial infarction, and/or a severe

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calcified aortic valve stenosis. For subjects that are genotyped or determined to be either LY75, CD164, and PARP1 reference or heterozygous for one or more LY75, CD164, or PARP1 variant nucleic acid molecules, such subject or subjects can be treated with an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof.

5 In any of the embodiments described herein, the subject in whom CHIP is prevented or reduced by administering an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, can be anyone at risk for developing CHIP including, but not limited to, subjects with CHIP-related disorders. In addition, in some embodiments, the subject is at risk of developing CHIP. In some embodiments, administering an LY75 inhibitor, a CD164 inhibitor, or a
10 PARP1 inhibitor, or any combination thereof, may be carried out to prevent development of an additional CHIP or CHIP-related disorders in a subject who has already had CHIP or a CHIP-related disorder.

In any of the embodiments described herein, the LY75 variant nucleic acid molecule can be any nucleic acid molecule (such as, for example, genomic nucleic acid molecule, mRNA
15 molecule, or cDNA molecule) encoding an LY75 variant polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function. In some embodiments, the LY75 variant nucleic acid molecules encoding an LY75 predicted loss-of-function polypeptide is associated with a reduced *in vitro* response to LY75 ligands compared with reference LY75. In some embodiments, the LY75
20 variant nucleic acid molecules encoding an LY75 predicted loss-of-function polypeptide is an LY75 variant that results or is predicted to result in a premature truncation of an LY75 polypeptide compared to the human reference genome sequence.

In any of the embodiments described herein, the CD164 variant nucleic acid molecule can be any nucleic acid molecule (such as, for example, genomic nucleic acid molecule, mRNA
25 molecule, or cDNA molecule) encoding a CD164 variant polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function. In some embodiments, the CD164 variant nucleic acid molecules encoding a CD164 predicted loss-of-function polypeptide is associated with a reduced *in vitro* response to CD164 ligands compared with reference CD164. In some embodiments, the CD164
30 variant nucleic acid molecules encoding a CD164 predicted loss-of-function polypeptide is a CD164 variant that results or is predicted to result in a premature truncation of a CD164 polypeptide compared to the human reference genome sequence.

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In any of the embodiments described herein, the PARP1 variant nucleic acid molecule can be any nucleic acid molecule (such as, for example, genomic nucleic acid molecule, mRNA molecule, or cDNA molecule) encoding a PARP1 variant polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function. In some embodiments, the PARP1 variant nucleic acid molecules encoding a PARP1 predicted loss-of-function polypeptide is associated with a reduced *in vitro* response to PARP1 ligands compared with reference PARP1. In some embodiments, the PARP1 variant nucleic acid molecules encoding a PARP1 predicted loss-of-function polypeptide is a PARP1 variant that results or is predicted to result in a premature truncation of a PARP1 polypeptide compared to the human reference genome sequence.

In some embodiments, the LY75, CD164, or PARP1 variant nucleic acid molecules are variants that are predicted to be damaging by *in vitro* prediction algorithms such as Polyphen, SIFT, or similar algorithms.

In some embodiments, the LY75 variant nucleic acid molecules are variants that cause or are predicted to cause a nonsynonymous amino-acid substitution in an LY75 nucleic acid molecule and whose allele frequency is less than 1/100 alleles in the population from which the subject is selected. In some embodiments, the LY75 variant nucleic acid molecule is any rare variant (allele frequency < 0.1%; or 1 in 1,000 alleles), or any splice-site, stop-gain, start-loss, stop-loss, frameshift, or in-frame indel, or other frameshift LY75 variant.

In any of the embodiments described herein, the LY75 predicted loss-of-function polypeptide can be any LY75 polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function.

In any of the embodiments described herein, the LY75 variant nucleic acid molecule can include variations at positions of chromosome 2 using the nucleotide sequence of the LY75 reference genomic nucleic acid molecule (SEQ ID NO:1; ENSG00000054219.11, chr2:159,803,355-159,904,756 or in the GRCh38/hg38 human genome assembly) as a reference sequence. Numerous genetic variants in LY75 exist which cause subsequent changes in the LY75 polypeptide sequence including, but not limited to rs78446341 (GRCh38/hg38 chr2:159,834,145:G:A) and rs147820690 (GRCh38/hg38 chr2:159,878,663:C:T).

In some embodiments, the CD164 variant nucleic acid molecules are variants that cause or are predicted to cause a nonsynonymous amino-acid substitution in CD164 nucleic acid molecules and whose allele frequency is less than 1/100 alleles in the population from

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which the subject is selected. In some embodiments, the CD164 variant nucleic acid molecules are any rare variant (allele frequency < 0.1%; or 1 in 1,000 alleles), or any splice-site, stop-gain, start-loss, stop-loss, frameshift, or in-frame indel, or other frameshift CD164 variant.

In any of the embodiments described herein, the CD164 predicted loss-of-function polypeptide can be any CD164 polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function.

In any of the embodiments described herein, the CD164 variant nucleic acid molecule can include variations at positions of chromosome 6 using the nucleotide sequence of the CD164 reference genomic nucleic acid molecule (SEQ ID NO:55; ENSG00000135535.17, chr6:109,366,514-109,381,739 in the GRCh38/hg38 human genome assembly) as a reference sequence. Numerous genetic variants in CD164 exist which cause subsequent changes in the CD164 polypeptide sequence including, but not limited to rs3799840 (GRCh38/hg38 chr6:109381443A:T).

In some embodiments, the PARP1 variant nucleic acid molecules are variants that cause or are predicted to cause a nonsynonymous amino-acid substitution in PARP1 nucleic acid molecules and whose allele frequency is less than 1/100 alleles in the population from which the subject is selected. In some embodiments, the PARP1 variant nucleic acid molecules are any rare variant (allele frequency < 0.1%; or 1 in 1,000 alleles), or any splice-site, stop-gain, start-loss, stop-loss, frameshift, or in-frame indel, or other frameshift PARP1 variant.

In any of the embodiments described herein, the PARP1 predicted loss-of-function polypeptide can be any PARP1 polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function.

In any of the embodiments described herein, the PARP1 variant nucleic acid molecule can include variations at positions of chromosome 1 using the nucleotide sequence of the PARP1 reference genomic nucleic acid molecule (SEQ ID NO:113; ENSG00000143799.14, chr1:226,360,691-226,408,093 in the GRCh38/hg38 human genome assembly) as a reference sequence. Numerous genetic variants in PARP1 exist which cause subsequent changes in the PARP1 polypeptide sequence including, but not limited to s1136410 (GRCh38/hg38 chr1:226367601:A:G), rs2793379 (GRCh38/hg38 chr1:226408985:T:A), rs2570367 (GRCh38/hg38 chr1:226414809T:C), rs1433574 (GRCh38/hg38 chr1:226421638:A:C) and rs2039925 (GRCh38/hg38chr1:226422811:C:G).

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Any one or more (i.e., any combination) of the LY75, CD164, or PARP1 variant nucleic acid molecules can be used within any of the methods described herein to determine whether a subject has an increased risk of developing CHIP or a CHIP-related disorder. The combinations of particular variants can form a mask used for statistical analysis of the particular correlation of any one or more LY75, CD164, or PARP1 and decreased risk of developing CHIP.

In any of the embodiments described herein, the CHIP or CHIP-related disorder is a hematologic cancer, a myeloid neoplasia, a lymphoid neoplasia, an atherosclerotic cardiovascular disease, a coronary heart disease, a myocardial infarction, and/or a severe calcified aortic valve stenosis. In some embodiments, the CHIP or CHIP-related disorder is a hematologic cancer. In some embodiments, the CHIP or CHIP-related disorder is a myeloid neoplasia. In some embodiments, the CHIP or CHIP-related disorder is a lymphoid neoplasia. In some embodiments, the CHIP or CHIP-related disorder is an atherosclerotic cardiovascular disease. In some embodiments, the CHIP or CHIP-related disorder is a coronary heart disease. In some embodiments, the CHIP or CHIP-related disorder is a myocardial infarction. In some embodiments, the CHIP or CHIP-related disorder is a severe calcified aortic valve stenosis.

Symptoms of myeloid neoplasia include, but are not limited to, fever, bone pain, lethargy and fatigue, shortness of breath, pale skin, frequent infections, easy bruising, and unusual bleeding, such as frequent nosebleeds and bleeding from the gums.

Symptoms of lymphoid neoplasia include, but are not limited to, painless swelling of lymph nodes in neck, armpits or groin, persistent fatigue, fever, night sweats, shortness of breath, unexplained weight loss, and itchy skin.

Symptoms of coronary heart disease include, but are not limited to, angina, cold sweats, dizziness, light-headedness, nausea or a feeling of indigestion, neck pain, shortness of breath (especially with activity), sleep disturbances, and weakness.

Symptoms of myocardial infarction include, but are not limited to, pressure or tightness in the chest, pain in the chest, back, jaw, and other areas of the upper body that lasts more than a few minutes or that goes away and comes back, shortness of breath, sweating, nausea, vomiting, anxiety, a cough, dizziness, and a fast heart rate.

Symptoms of severe calcified aortic valve stenosis include, but are not limited to, abnormal heart sound (heart murmur) heard through a stethoscope, chest pain (angina) or tightness (with activity), feeling faint or dizzy or fainting (with activity), shortness of breath,

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(especially with activity), fatigue (especially during times of increased activity), rapid, fluttering heartbeat (palpitations), not eating enough (mainly in children with aortic valve stenosis), and not gaining enough weight (mainly in children with aortic valve stenosis).

It has also been observed in accordance with the present disclosure that CHIP somatic mutations in either DNMT3A or ASXL1 associate with an increased risk of developing lung cancer. Therefore, subjects that have a CHIP somatic mutation in either DNMT3A or ASXL1 may be monitored more frequently for lung cancer pathology (such as by more frequent chest x-rays, or the like), treatment with palliative agents, or smoking cessation procedures, such that development of lung cancer is prevented or delayed.

In any of the embodiments described herein, a DNMT3A or an ASXL1 somatic mutation is any mutation that is a missense mutation, a splice-site mutation, a stop-gain mutation, a start-loss mutation, a stop-loss mutation, a frameshift mutation, an in-frame indel mutation, or a mutation that results in a truncated DNMT3A or ASXL1 polypeptide. In any of the embodiments described herein, a DNMT3A or an ASXL1 somatic mutation can also be any mutation that results in a DNMT3A or an ASXL1 polypeptide having a partial loss-of-function, a complete loss-of-function, a predicted partial loss-of-function, or a predicted complete loss-of-function.

In any of the embodiments described herein, the DNMT3A mutation can include variations at positions of chromosome 2 using the nucleotide sequence of the DNMT3A reference genomic nucleic acid molecule (SEQ ID NO:212; ENSG00000119772.17, chr2:25,227,855-25,342,590 in the GRCh38/hg38 human genome assembly) as a reference sequence.

In any of the embodiments described herein, the ASXL1 mutation can include variations at positions of chromosome 20 using the nucleotide sequence of the ASXL1 reference genomic nucleic acid molecule (SEQ ID NO:267; ENSG00000171456.20, chr20:32,358,330-32,439,260 in the GRCh38/hg38 human genome assembly) as a reference sequence.

The present disclosure provides methods of preventing or reducing the development of CHIP in a subject, the methods comprising administering an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject.

The present disclosure also provides methods of treating a subject having a hematologic cancer or at risk of developing a hematologic cancer, the methods comprising

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administering an LY75 inhibitor, or a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject.

The present disclosure also provides methods of treating a subject having a myeloid neoplasia or at risk of developing a myeloid neoplasia, the methods comprising administering
5 an LY75 inhibitor, or a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject.

The present disclosure also provides methods of treating a subject having a lymphoid neoplasia or at risk of developing a lymphoid neoplasia, the methods comprising administering
10 an LY75 inhibitor, or a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject.

The present disclosure also provides methods of treating a subject having an atherosclerotic cardiovascular disease or at risk of developing an atherosclerotic cardiovascular disease, the methods comprising administering an LY75 inhibitor, or a CD164 inhibitor, or a
PARP1 inhibitor, or any combination thereof, to the subject.

15 The present disclosure also provides methods of treating a subject having a coronary heart disease or at risk of developing a coronary heart disease, the methods comprising administering an LY75 inhibitor, or a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject.

The present disclosure also provides methods of treating a subject who has or has had
20 a myocardial infarction or at risk of developing a myocardial infarction, the methods comprising administering an LY75 inhibitor, or a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject.

The present disclosure also provides methods of treating a subject having a severe calcified aortic valve stenosis or at risk of developing a severe calcified aortic valve stenosis, the
25 methods comprising administering an LY75 inhibitor, or a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject.

In some embodiments, the LY75 inhibitor comprises an inhibitory nucleic acid molecule. Examples of inhibitory nucleic acid molecules include, but are not limited to, antisense nucleic acid molecules, small interfering RNAs (siRNAs), and short hairpin RNAs
30 (shRNAs). Such inhibitory nucleic acid molecules can be designed to target any region of an LY75 nucleic acid molecule. In some embodiments, the antisense RNA, siRNA, or shRNA hybridizes to a sequence within an LY75 genomic nucleic acid molecule or mRNA molecule and

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decreases expression of the LY75 polypeptide in a cell in the subject. In some embodiments, the LY75 inhibitor comprises an antisense molecule that hybridizes to an LY75 genomic nucleic acid molecule or mRNA molecule and decreases expression of the LY75 polypeptide in a cell in the subject. In some embodiments, the LY75 inhibitor comprises an siRNA that hybridizes to an
5 LY75 genomic nucleic acid molecule or mRNA molecule and decreases expression of the LY75 polypeptide in a cell in the subject. In some embodiments, the LY75 inhibitor comprises an shRNA that hybridizes to an LY75 genomic nucleic acid molecule or mRNA molecule and decreases expression of the LY75 polypeptide in a cell in the subject.

In some embodiments, the CD164 inhibitor comprises an inhibitory nucleic acid
10 molecule. Examples of inhibitory nucleic acid molecules include, but are not limited to, antisense nucleic acid molecules, small interfering RNAs (siRNAs), and short hairpin RNAs (shRNAs). Such inhibitory nucleic acid molecules can be designed to target any region of a CD164 nucleic acid molecule. In some embodiments, the antisense RNA, siRNA, or shRNA hybridizes to a sequence within a CD164 genomic nucleic acid molecule or mRNA molecule and
15 decreases expression of the CD164 polypeptide in a cell in the subject. In some embodiments, the CD164 inhibitor comprises an antisense molecule that hybridizes to a CD164 genomic nucleic acid molecule or mRNA molecule and decreases expression of the CD164 polypeptide in a cell in the subject. In some embodiments, the CD164 inhibitor comprises an siRNA that hybridizes to a CD164 genomic nucleic acid molecule or mRNA molecule and decreases
20 expression of the CD164 polypeptide in a cell in the subject. In some embodiments, the CD164 inhibitor comprises an shRNA that hybridizes to a CD164 genomic nucleic acid molecule or mRNA molecule and decreases expression of the CD164 polypeptide in a cell in the subject.

In some embodiments, the PARP1 inhibitor comprises an inhibitory nucleic acid molecule. Examples of inhibitory nucleic acid molecules include, but are not limited to,
25 antisense nucleic acid molecules, small interfering RNAs (siRNAs), and short hairpin RNAs (shRNAs). Such inhibitory nucleic acid molecules can be designed to target any region of a PARP1 nucleic acid molecule. In some embodiments, the antisense RNA, siRNA, or shRNA hybridizes to a sequence within a PARP1 genomic nucleic acid molecule or mRNA molecule and decreases expression of the PARP1 polypeptide in a cell in the subject. In some embodiments,
30 the PARP1 inhibitor comprises an antisense molecule that hybridizes to a PARP1 genomic nucleic acid molecule or mRNA molecule and decreases expression of the PARP1 polypeptide in a cell in the subject. In some embodiments, the PARP1 inhibitor comprises an siRNA that

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hybridizes to a PARP1 genomic nucleic acid molecule or mRNA molecule and decreases expression of the PARP1 polypeptide in a cell in the subject. In some embodiments, the PARP1 inhibitor comprises an shRNA that hybridizes to a PARP1 genomic nucleic acid molecule or mRNA molecule and decreases expression of the PARP1 polypeptide in a cell in the subject.

5 The inhibitory nucleic acid molecules can comprise RNA, DNA, or both RNA and DNA. The inhibitory nucleic acid molecules can also be linked or fused to a heterologous nucleic acid sequence, such as in a vector, or a heterologous label. For example, the inhibitory nucleic acid molecules can be within a vector or as an exogenous donor sequence comprising the inhibitory nucleic acid molecule and a heterologous nucleic acid sequence. The inhibitory nucleic acid
10 molecules can also be linked or fused to a heterologous label. The label can be directly detectable (such as, for example, fluorophore) or indirectly detectable (such as, for example, hapten, enzyme, or fluorophore quencher). Such labels can be detectable by spectroscopic, photochemical, biochemical, immunochemical, or chemical means. Such labels include, for example, radiolabels, pigments, dyes, chromogens, spin labels, and fluorescent labels. The label
15 can also be, for example, a chemiluminescent substance; a metal-containing substance; or an enzyme, where there occurs an enzyme-dependent secondary generation of signal. The term "label" can also refer to a "tag" or hapten that can bind selectively to a conjugated molecule such that the conjugated molecule, when added subsequently along with a substrate, is used to generate a detectable signal. For example, biotin can be used as a tag along with an avidin or
20 streptavidin conjugate of horseradish peroxidase (HRP) to bind to the tag, and examined using a calorimetric substrate (such as, for example, tetramethylbenzidine (TMB)) or a fluorogenic substrate to detect the presence of HRP. Exemplary labels that can be used as tags to facilitate purification include, but are not limited to, myc, HA, FLAG or 3XFLAG, 6XHis or polyhistidine, glutathione-S-transferase (GST), maltose binding protein, an epitope tag, or the Fc portion of
25 immunoglobulin. Numerous labels include, for example, particles, fluorophores, haptens, enzymes and their calorimetric, fluorogenic and chemiluminescent substrates and other labels.

In any of the embodiments described herein, any of the inhibitory nucleic acid molecules can be formulated as a component of a lipid nanoparticle, and can be delivered to a cell by a lipid nanoparticle.

30 The inhibitory nucleic acid molecules can comprise, for example, nucleotides or non-natural or modified nucleotides, such as nucleotide analogs or nucleotide substitutes. Such nucleotides include a nucleotide that contains a modified base, sugar, or phosphate group, or

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that incorporates a non-natural moiety in its structure. Examples of non-natural nucleotides include, but are not limited to, dideoxynucleotides, biotinylated, aminated, deaminated, alkylated, benzylated, and fluorophor-labeled nucleotides.

The inhibitory nucleic acid molecules can also comprise one or more nucleotide
5 analogs or substitutions. A nucleotide analog is a nucleotide which contains a modification to either the base, sugar, or phosphate moieties. Modifications to the base moiety include, but are not limited to, natural and synthetic modifications of A, C, G, and T/U, as well as different purine or pyrimidine bases such as, for example, pseudouridine, uracil-5-yl, hypoxanthin-9-yl (I), and 2-aminoadenin-9-yl. Modified bases include, but are not limited to, 5-methylcytosine
10 (5-me-C), 5-hydroxymethyl cytosine, xanthine, hypoxanthine, 2-aminoadenine, 6-methyl and other alkyl derivatives of adenine and guanine, 2-propyl and other alkyl derivatives of adenine and guanine, 2-thiouracil, 2-thiothymine and 2-thiocytosine, 5-halouracil and cytosine, 5-propynyl uracil and cytosine, 6-azo uracil, cytosine and thymine, 5-uracil (pseudouracil), 4-thiouracil, 8-halo, 8-amino, 8-thiol, 8-thioalkyl, 8-hydroxyl and other 8-substituted adenines
15 and guanines, 5-halo (such as, for example, 5-bromo), 5-trifluoromethyl and other 5-substituted uracils and cytosines, 7-methylguanine, 7-methyladenine, 8-azaguanine, 8-azaadenine, 7-deazaguanine, 7-deazaadenine, 3-deazaguanine, and 3-deazaadenine.

Nucleotide analogs can also include modifications of the sugar moiety. Modifications to the sugar moiety include, but are not limited to, natural modifications of the ribose and
20 deoxy ribose as well as synthetic modifications. Sugar modifications include, but are not limited to, the following modifications at the 2' position: OH; F; O-, S-, or N-alkyl; O-, S-, or N-alkenyl; O-, S- or N-alkynyl; or O-alkyl-O-alkyl, wherein the alkyl, alkenyl, and alkynyl may be substituted or unsubstituted C₁₋₁₀alkyl or C₂₋₁₀alkenyl, and C₂₋₁₀alkynyl. Exemplary 2' sugar modifications also include, but are not limited to, -O[(CH₂)_nO]_mCH₃, -O(CH₂)_nOCH₃, -O(CH₂)_nNH₂, -O(CH₂)_nCH₃,
25 -O(CH₂)_n-ONH₂, and -O(CH₂)_nON[(CH₂)_nCH₃]₂, where n and m, independently, are from 1 to about 10. Other modifications at the 2' position include, but are not limited to, C₁₋₁₀alkyl, substituted lower alkyl, alkaryl, aralkyl, O-alkaryl or O-aralkyl, SH, SCH₃, OCN, Cl, Br, CN, CF₃, OCF₃, SOCH₃, SO₂CH₃, ONO₂, NO₂, N₃, NH₂, heterocycloalkyl, heterocycloalkaryl, aminoalkylamino, polyalkylamino, substituted silyl, an RNA cleaving group, a reporter group, an
30 intercalator, a group for improving the pharmacokinetic properties of an oligonucleotide, or a group for improving the pharmacodynamic properties of an oligonucleotide, and other substituents having similar properties. Similar modifications may also be made at other

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positions on the sugar, particularly the 3' position of the sugar on the 3' terminal nucleotide or in 2'-5' linked oligonucleotides and the 5' position of 5' terminal nucleotide. Modified sugars can also include those that contain modifications at the bridging ring oxygen, such as CH₂ and S. Nucleotide sugar analogs can also have sugar mimetics, such as cyclobutyl moieties in place of the pentofuranosyl sugar.

Nucleotide analogs can also be modified at the phosphate moiety. Modified phosphate moieties include, but are not limited to, those that can be modified so that the linkage between two nucleotides contains a phosphorothioate, chiral phosphorothioate, phosphorodithioate, phosphotriester, aminoalkylphosphotriester, methyl and other alkyl phosphonates including 3'-alkylene phosphonate and chiral phosphonates, phosphinates, phosphoramidates including 3'-amino phosphoramidate and aminoalkylphosphoramidates, thionophosphoramidates, thionoalkylphosphonates, thionoalkylphosphotriesters, and boranophosphates. These phosphate or modified phosphate linkage between two nucleotides can be through a 3'-5' linkage or a 2'-5' linkage, and the linkage can contain inverted polarity such as 3'-5' to 5'-3' or 2'-5' to 5'-2'. Various salts, mixed salts, and free acid forms are also included. Nucleotide substitutes also include peptide nucleic acids (PNAs).

In some embodiments, the antisense nucleic acid molecules are gapmers, whereby the first one to seven nucleotides at the 5' and 3' ends each have 2'-methoxyethyl (2'-MOE) modifications. In some embodiments, the first five nucleotides at the 5' and 3' ends each have 2'-MOE modifications. In some embodiments, the first one to seven nucleotides at the 5' and 3' ends are RNA nucleotides. In some embodiments, the first five nucleotides at the 5' and 3' ends are RNA nucleotides. In some embodiments, each of the backbone linkages between the nucleotides is a phosphorothioate linkage.

In some embodiments, the siRNA molecules have termini modifications. In some embodiments, the 5' end of the antisense strand is phosphorylated. In some embodiments, 5'-phosphate analogs that cannot be hydrolyzed, such as 5'-(E)-vinyl-phosphonate are used.

In some embodiments, the siRNA molecules have backbone modifications. In some embodiments, the modified phosphodiester groups that link consecutive ribose nucleosides have been shown to enhance the stability and *in vivo* bioavailability of siRNAs. The non-ester groups (-OH, =O) of the phosphodiester linkage can be replaced with sulfur, boron, or acetate to give phosphorothioate, boranophosphate, and phosphonoacetate linkages. In addition, substituting the phosphodiester group with a phosphotriester can facilitate cellular uptake of

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siRNAs and retention on serum components by eliminating their negative charge. In some embodiments, the siRNA molecules have sugar modifications. In some embodiments, the sugars are deprotonated (reaction catalyzed by exo- and endonucleases) whereby the 2'-hydroxyl can act as a nucleophile and attack the adjacent phosphorous in the phosphodiester bond. Such alternatives include 2'-O-methyl, 2'-O-methoxyethyl, and 2'-fluoro modifications.

In some embodiments, the siRNA molecules have base modifications. In some embodiments, the bases can be substituted with modified bases such as pseudouridine, 5'-methylcytidine, N6-methyladenosine, inosine, and N7-methylguanosine.

In some embodiments, the siRNA molecules are conjugated to lipids. Lipids can be conjugated to the 5' or 3' termini of siRNA to improve their *in vivo* bioavailability by allowing them to associate with serum lipoproteins. Representative lipids include, but are not limited to, cholesterol and vitamin E, and fatty acids, such as palmitate and tocopherol.

In some embodiments, a representative siRNA has the following formula:

Sense: mN*mN*/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/

i2FN/*mN*/32FN/

Antisense: /52FN*/i2FN/*mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN/i2FN/mN*N*N

wherein: "N" is the base; "2F" is a 2'-F modification; "m" is a 2'-O-methyl modification, "I" is an internal base; and "*" is a phosphorothioate backbone linkage.

The present disclosure also provides vectors comprising any one or more of the inhibitory nucleic acid molecules. In some embodiments, the vectors comprise any one or more of the inhibitory nucleic acid molecules and a heterologous nucleic acid. The vectors can be viral or nonviral vectors capable of transporting a nucleic acid molecule. In some embodiments, the vector is a plasmid or cosmid (such as, for example, a circular double-stranded DNA into which additional DNA segments can be ligated). In some embodiments, the vector is a viral vector, wherein additional DNA segments can be ligated into the viral genome. Expression vectors include, but are not limited to, plasmids, cosmids, retroviruses, adenoviruses, adeno-associated viruses (AAV), plant viruses such as cauliflower mosaic virus and tobacco mosaic virus, yeast artificial chromosomes (YACs), Epstein-Barr (EBV)-derived episomes, and other expression vectors known in the art.

The present disclosure also provides compositions comprising any one or more of the inhibitory nucleic acid molecules. In some embodiments, the composition is a pharmaceutical

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composition. In some embodiments, the compositions comprise a carrier and/or excipient. Examples of carriers include, but are not limited to, poly(lactic acid) (PLA) microspheres, poly(D,L-lactic-coglycolic-acid) (PLGA) microspheres, liposomes, micelles, inverse micelles, lipid cochleates, and lipid microtubules. A carrier may comprise a buffered salt solution such as PBS, HBSS, etc.

In some embodiments, the LY75 inhibitor, the CD164 inhibitor, or the PARP1 inhibitor comprises a nuclease agent that induces one or more nicks or double-strand breaks at a recognition sequence(s) or a DNA-binding protein that binds to a recognition sequence within an LY75, a CD164, or a PARP1 genomic nucleic acid molecule. The recognition sequence can be located within a coding region of the LY75 gene, the CD164 gene, or the PARP1 gene, or within regulatory regions that influence the expression of the gene. A recognition sequence of the DNA-binding protein or nuclease agent can be located in an intron, an exon, a promoter, an enhancer, a regulatory region, or any non-protein coding region. The recognition sequence can include or be proximate to the start codon of the LY75 gene, the CD164 gene, or the PARP1 gene. For example, the recognition sequence can be located about 10, about 20, about 30, about 40, about 50, about 100, about 200, about 300, about 400, about 500, or about 1,000 nucleotides from the start codon. As another example, two or more nuclease agents can be used, each targeting a nuclease recognition sequence including or proximate to the start codon. As another example, two nuclease agents can be used, one targeting a nuclease recognition sequence including or proximate to the start codon, and one targeting a nuclease recognition sequence including or proximate to the stop codon, wherein cleavage by the nuclease agents can result in deletion of the coding region between the two nuclease recognition sequences. Any nuclease agent that induces a nick or double-strand break into a desired recognition sequence can be used in the methods and compositions disclosed herein. Any DNA-binding protein that binds to a desired recognition sequence can be used in the methods and compositions disclosed herein.

Suitable nuclease agents and DNA-binding proteins for use herein include, but are not limited to, zinc finger protein or zinc finger nuclease (ZFN) pair, Transcription Activator-Like Effector (TALE) protein or Transcription Activator-Like Effector Nuclease (TALEN), or Clustered Regularly Interspersed Short Palindromic Repeats (CRISPR)/CRISPR-associated (Cas) systems. The length of the recognition sequence can vary, and includes, for example, recognition sequences that are about 30-36 bp for a zinc finger protein or ZFN pair, about 15-18 bp for each

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ZFN, about 36 bp for a TALE protein or TALEN, and about 20 bp for a CRISPR/Cas guide RNA.

In some embodiments, CRISPR/Cas systems can be used to modify LY75, CD164, and/or PARP1 genomic nucleic acid molecule within a cell. The methods and compositions disclosed herein can employ CRISPR-Cas systems by utilizing CRISPR complexes (comprising a guide RNA (gRNA) complexed with a Cas protein) for site-directed cleavage of LY75, CD164, and/or PARP1 nucleic acid molecules.

Cas proteins generally comprise at least one RNA recognition or binding domain that can interact with gRNAs. Cas proteins can also comprise nuclease domains (such as, for example, DNase or RNase domains), DNA binding domains, helicase domains, protein-protein interaction domains, dimerization domains, and other domains. Suitable Cas proteins include, for example, a wild type Cas9 protein and a wild type Cpf1 protein (such as, for example, FnCpf1). A Cas protein can have full cleavage activity to create a double-strand break in an LY75, a CD164, or a PARP1 genomic nucleic acid molecule or it can be a nickase that creates a single-strand break in an LY75, a CD164, or a PARP1 genomic nucleic acid molecule. Additional examples of Cas proteins include, but are not limited to, Cas1, Cas1B, Cas2, Cas3, Cas4, Cas5, Cas5e (CasD), Cas6, Cas6e, Cas6f, Cas7, Cas8a1, Cas8a2, Cas8b, Cas8c, Cas9 (Csn1 or Csx12), Cas10, Cas10d, CasF, CasG, CasH, Csy1, Csy2, Csy3, Cse1 (CasA), Cse2 (CasB), Cse3 (CasE), Cse4 (CasC), Csc1, Csc2, Csa5, Csn2, Csm2, Csm3, Csm4, Csm5, Csm6, Cmr1, Cmr3, Cmr4, Cmr5, Cmr6, Csb1, Csb2, Csb3, Csx17, Csx14, Csx10, Csx16, CsaX, Csx3, Csx1, Csx15, Csf1, Csf2, Csf3, Csf4, and Cu1966, and homologs or modified versions thereof. Cas proteins can also be operably linked to heterologous polypeptides as fusion proteins. For example, a Cas protein can be fused to a cleavage domain, an epigenetic modification domain, a transcriptional activation domain, or a transcriptional repressor domain. Cas proteins can be provided in any form. For example, a Cas protein can be provided in the form of a protein, such as a Cas protein complexed with a gRNA. Alternately, a Cas protein can be provided in the form of a nucleic acid molecule encoding the Cas protein, such as an RNA or DNA.

In some embodiments, targeted genetic modifications of LY75 genomic nucleic acid molecules can be generated by contacting a cell with a Cas protein and one or more gRNAs that hybridize to one or more gRNA recognition sequences within a target genomic locus in the LY75 genomic nucleic acid molecule. For example, an LY75 gRNA recognition sequence can be located within a region of SEQ ID NO:1. The gRNA recognition sequence can include or be proximate to the start codon of an LY75 genomic nucleic acid molecule or the stop codon of an

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LY75 genomic nucleic acid molecule. For example, the gRNA recognition sequence can be located from about 10, from about 20, from about 30, from about 40, from about 50, from about 100, from about 200, from about 300, from about 400, from about 500, or from about 1,000 nucleotides of the start codon or the stop codon.

5 In some embodiments, targeted genetic modifications of CD164 genomic nucleic acid molecules can be generated by contacting a cell with a Cas protein and one or more gRNAs that hybridize to one or more gRNA recognition sequences within a target genomic locus in the CD164 genomic nucleic acid molecule. For example, a CD164 gRNA recognition sequence can be located within a region of SEQ ID NO:55. The gRNA recognition sequence can include or be
10 proximate to the start codon of a CD164 genomic nucleic acid molecule or the stop codon of a CD164 genomic nucleic acid molecule. For example, the gRNA recognition sequence can be located from about 10, from about 20, from about 30, from about 40, from about 50, from about 100, from about 200, from about 300, from about 400, from about 500, or from about 1,000 nucleotides of the start codon or the stop codon.

15 In some embodiments, targeted genetic modifications of PARP1 genomic nucleic acid molecules can be generated by contacting a cell with a Cas protein and one or more gRNAs that hybridize to one or more gRNA recognition sequences within a target genomic locus in the PARP1 genomic nucleic acid molecule. For example, a PARP1 gRNA recognition sequence can be located within a region of SEQ ID NO:113. The gRNA recognition sequence can include or be
20 proximate to the start codon of a PARP1 genomic nucleic acid molecule or the stop codon of a PARP1 genomic nucleic acid molecule. For example, the gRNA recognition sequence can be located from about 10, from about 20, from about 30, from about 40, from about 50, from about 100, from about 200, from about 300, from about 400, from about 500, or from about 1,000 nucleotides of the start codon or the stop codon.

25 The gRNA recognition sequences within a target genomic locus in an LY75, a CD164, or a PARP1 genomic nucleic acid molecule are located near a Protospacer Adjacent Motif (PAM) sequence, which is a 2-6 base pair DNA sequence immediately following the DNA sequence targeted by the Cas9 nuclease. The canonical PAM is the sequence 5'-NGG-3' where "N" is any nucleobase followed by two guanine ("G") nucleobases. gRNAs can transport Cas9 to anywhere
30 in the genome for gene editing, but no editing can occur at any site other than one at which Cas9 recognizes PAM. In addition, 5'-NGA-3' can be a highly efficient non-canonical PAM for human cells. Generally, the PAM is about 2-6 nucleotides downstream of the DNA sequence

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targeted by the gRNA. The PAM can flank the gRNA recognition sequence. In some embodiments, the gRNA recognition sequence can be flanked on the 3' end by the PAM. In some embodiments, the gRNA recognition sequence can be flanked on the 5' end by the PAM. For example, the cleavage site of Cas proteins can be about 1 to about 10, about 2 to about 5
5 base pairs, or three base pairs upstream or downstream of the PAM sequence. In some embodiments (such as when Cas9 from *S. pyogenes* or a closely related Cas9 is used), the PAM sequence of the non-complementary strand can be 5'-NGG-3', where N is any DNA nucleotide and is immediately 3' of the gRNA recognition sequence of the non-complementary strand of the target DNA. As such, the PAM sequence of the complementary strand would be 5'-CCN-3',
10 where N is any DNA nucleotide and is immediately 5' of the gRNA recognition sequence of the complementary strand of the target DNA.

A gRNA is an RNA molecule that binds to a Cas protein and targets the Cas protein to a specific location within an LY75, a CD164, or a PARP1 genomic nucleic acid molecule. An exemplary gRNA is a gRNA effective to direct a Cas enzyme to bind to or cleave an LY75, a
15 CD164, or a PARP1 genomic nucleic acid molecule, wherein the gRNA comprises a DNA-targeting segment that hybridizes to a gRNA recognition sequence within the LY75, CD164, or PARP1 genomic nucleic acid molecule. Exemplary gRNAs comprise a DNA-targeting segment that hybridizes to a gRNA recognition sequence present within an LY75, a CD164, or a PARP1 genomic nucleic acid molecule that includes or is proximate to the start codon or the stop
20 codon. For example, a gRNA can be selected such that it hybridizes to a gRNA recognition sequence that is located from about 5, from about 10, from about 15, from about 20, from about 25, from about 30, from about 35, from about 40, from about 45, from about 50, from about 100, from about 200, from about 300, from about 400, from about 500, or from about 1,000 nucleotides of the start codon or located from about 5, from about 10, from about 15,
25 from about 20, from about 25, from about 30, from about 35, from about 40, from about 45, from about 50, from about 100, from about 200, from about 300, from about 400, from about 500, or from about 1,000 nucleotides of the stop codon. Suitable gRNAs can comprise from about 17 to about 25 nucleotides, from about 17 to about 23 nucleotides, from about 18 to about 22 nucleotides, or from about 19 to about 21 nucleotides. In some embodiments, the
30 gRNAs can comprise 20 nucleotides.

Examples of suitable gRNA recognition sequences located within the human LY75 reference gene are set forth in Table 1 as SEQ ID NOs:152-171.

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Table 1: Guide RNA Recognition Sequences Near LY75

Strand	gRNA Recognition Sequence	SEQ ID NO:
-	GGTGGATAAGAATTAGCGAG	152
-	GTGGAAAGTCCAATCATGTG	153
-	TATGAATATGACCGAAAGTG	154
-	GAGTATAACTGGGCAACTGT	155
-	TGTGTAGTATTAGACACTGA	156
-	TGTGAGCACCCTCTCTGTA	157
+	GAATAGTAGCAAATAGCACC	158
-	GAAACCATTAAATAATACAG	159
+	GGTTGAGTATTAGGGCACAG	160
-	GTGTTGAGCCAGATTGAACA	161
-	AATCACCGTTTACTGGGACG	162
-	ACACCAGACTGGTACAATCC	163
+	ACAGGGGTCATCAGGCTTAG	164
-	GCTATACTCTGCTAGAGGCT	165
+	CAGGTCCCATCAATTAAGAA	166
-	AATCAGCTATACTCTGCTAG	167
+	CAGCTGGAGCCACCTATAGT	168
+	TTCACAGATGAAGGGCAACT	169
+	GCCAGGATCCTTGTAATCT	170
+	CTCACATTTCCACCACAGCA	171

Examples of suitable gRNA recognition sequences located within the human CD164 reference gene are set forth in Table 2 as SEQ ID NOs:172-191.

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Table 2: Guide RNA Recognition Sequences Near CD164

Strand	gRNA Recognition Sequence	SEQ ID NO:
+	CTGGGTCGTGTTCTTGTCGG	172
-	AACTCCCCAGAAACCTGTGA	173
+	CACCGGCGCCGAGGTTACGT	174
-	ACAGTTAGTGATTGTCAAGT	175
-	GCCCATCTCCAACGTAACCT	176
-	CAGTTAGTGATTGTCAAGTG	177
+	ACCAGCGGGAGGGACGTCAC	178
+	GGTGCCGGAGTGGTGACCAG	179
-	CTCCAACGTAACCTCGGCGC	180
-	GTGCCAACAGCCAATTCTAC	181
-	CCTCCCGCTGGTCACCACTC	182
+	TCGACCTTCACAGTTTCTG	183
-	AACAGTTAGTGATTGTCAAG	184
+	GCCGAGGTTACGTTGGAGAT	185
+	GCAGCTGTTTCGACCTTCAC	186

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+	GAGGGACGTCACCGCGCCG	187
+	AGTCGTCACGTTCGGGTGCT	188
+	AACAACGCTAACATTAAC	189
+	GCAGAGCACGCCAGGCAGG	190
+	TTTACATTCTATCCAAAAGC	191

Examples of suitable gRNA recognition sequences located within the human PARP1 reference gene are set forth in Table 3 as SEQ ID NOs:192-211.

Table 3: Guide RNA Recognition Sequences Near PARP

Strand	gRNA Recognition Sequence	SEQ ID NO:
+	TACCGATCACCGTACCCACA	192
-	AACTCGGGGGAAGTTGACG	193
-	CGATGCCTATTACTGCACTG	194
-	AGCTAGGCATGATTGACCGC	195
+	TGGCCATAGTCAATCTCCAG	196
-	AATTATATGAAGAAAAACC	197
-	ATCTTGACCGAGTAGCTGA	198
-	GAAGGAGTGGGTAACCCCAA	199
+	GGCCATAGTCAATCTCCAGG	200
+	CTGGCCATAGTCAATCTCCA	201
-	GGAACAGATGCCGTCCAAGG	202
-	GACACAGACACCAACCGGA	203
+	GGAAGTCCTCAGACACAAC	204
-	GGCCATGATTGAGAACTCG	205
-	TCCAACAGAAGTACGTGCAA	206
-	TGGCCCCAAGAGGGAAGTCA	207
-	CATCCGGCACCTGACGTTG	208
-	CCCAGAAACAGCGCCTCCG	209
-	AAAGAGAAAAGCGATGAGG	210
-	CCTGGAGATTGACTATGGCC	211

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The Cas protein and the gRNA form a complex, and the Cas protein cleaves the target LY75, CD164, or PARP1 genomic nucleic acid molecule. The Cas protein can cleave the nucleic acid molecule at a site within or outside of the nucleic acid sequence present in the target LY75, CD164, or PARP1 genomic nucleic acid molecule to which the DNA-targeting segment of a gRNA will bind. For example, formation of a CRISPR complex (comprising a gRNA hybridized to a gRNA recognition sequence and complexed with a Cas protein) can result in cleavage of one or both strands in or near (such as, for example, within 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 20, 50, or more base

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pairs from) the nucleic acid sequence present in the LY75, CD164, or PARP1 genomic nucleic acid molecule to which a DNA-targeting segment of a gRNA will bind.

Such methods can result, for example, in an LY75 genomic nucleic acid molecule in which a region of SEQ ID NO:1 is disrupted, the start codon is disrupted, the stop codon is disrupted, or the coding sequence is disrupted or deleted. Optionally, the cell can be further contacted with one or more additional gRNAs that hybridize to additional gRNA recognition sequences within the target genomic locus in the LY75 genomic nucleic acid molecule. By contacting the cell with one or more additional gRNAs (such as, for example, a second gRNA that hybridizes to a second gRNA recognition sequence), cleavage by the Cas protein can create two or more double-strand breaks or two or more single-strand breaks. The CD164 and the PARP1 genomic DNAs can be similarly targeted.

In some embodiments, the methods of prevention and/or reduction further comprise detecting the presence or absence of an LY75 variant nucleic acid molecule in a biological sample from the subject. In some embodiments, the methods of prevention and/or reduction further comprise detecting the presence or absence of a CD164 variant nucleic acid molecule in a biological sample from the subject. In some embodiments, the methods of prevention and/or reduction further comprise detecting the presence or absence of a PARP1 variant nucleic acid molecule in a biological sample from the subject.

The present disclosure also provides methods of treating a subject with a therapeutic agent that treats or inhibits CHIP, wherein the subject is at risk of developing CHIP or a CHIP-related disorder. In some embodiments, the methods comprise determining whether the subject has an LY75 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has a genotype comprising an LY75 variant nucleic acid molecule. In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount to a subject that is LY75, and/or administering an LY75 inhibitor, to the subject. In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is heterozygous for the LY75 variant nucleic acid molecule, and/or administering an LY75 inhibitor. In some embodiments, the methods further comprise administering or continuing to

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administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is heterozygous for the LY75 variant nucleic acid molecule, and/or administering an LY75 inhibitor to the subject.

5 The present disclosure also provides methods of treating a subject with a therapeutic agent that treats or inhibits CHIP, wherein the subject is at risk of developing CHIP or a CHIP-related disorder. In some embodiments, the methods comprise determining whether the subject has a CD164 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has a genotype comprising a CD164 variant nucleic acid molecule. In some embodiments, the methods further comprise administering or
10 continuing to administer the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount to a subject that is CD164, and/or administering a CD164 inhibitor, to the subject. In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP
15 in an amount that is the same as or less than a standard dosage amount to a subject that is heterozygous for the CD164 variant nucleic acid molecule, and/or administering a CD164 inhibitor. In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is heterozygous for
20 the CD164 variant nucleic acid molecule, and/or administering a CD164 inhibitor to the subject.

The present disclosure also provides methods of treating a subject with a therapeutic agent that treats or inhibits CHIP, wherein the subject is at risk of developing CHIP or a CHIP-related disorder. In some embodiments, the methods comprise determining whether the subject has a PARP1 variant nucleic acid molecule by obtaining or having obtained a biological
25 sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has a genotype comprising a PARP1 variant nucleic acid molecule. In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount to a subject that is PARP1, and/or administering a PARP1
30 inhibitor, to the subject. In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is

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heterozygous for the PARP1 variant nucleic acid molecule, and/or administering a PARP1 inhibitor. In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is heterozygous for the PARP1 variant nucleic acid molecule, and/or administering a PARP1 inhibitor to the subject.

In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is homozygous for an LY75 variant nucleic acid molecule. The presence of a genotype having an LY75 variant nucleic acid molecule indicates the subject has a decreased risk of developing CHIP or a CHIP-related disorder. In some embodiments, the subject is LY75 reference. In some embodiments, the subject is heterozygous for an LY75 variant nucleic acid molecule.

In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is homozygous for a CD164 variant nucleic acid molecule. The presence of a genotype having a CD164 variant nucleic acid molecule indicates the subject has a decreased risk of developing CHIP or a CHIP-related disorder. In some embodiments, the subject is CD164 reference. In some embodiments, the subject is heterozygous for a CD164 variant nucleic acid molecule.

In some embodiments, the methods further comprise administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is homozygous for a PARP1 variant nucleic acid molecule. The presence of a genotype having a PARP1 variant nucleic acid molecule indicates the subject has a decreased risk of developing CHIP or a CHIP-related disorder. In some embodiments, the subject is PARP1 reference. In some embodiments, the subject is heterozygous for a PARP1 variant nucleic acid molecule.

For subjects that are genotyped or determined to be either LY75 reference or heterozygous for LY75 variant nucleic acid molecule, such subjects can be administered an LY75 inhibitor, as described herein.

For subjects that are genotyped or determined to be CD164 reference or heterozygous for CD164 variant nucleic acid molecule, such subjects can be administered a CD164 inhibitor, as described herein.

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For subjects that are genotyped or determined to be PARP1 reference or heterozygous for PARP1 variant nucleic acid molecule, such subjects can be administered a PARP1 inhibitor, as described herein.

For subjects that are genotyped or determined to be either or both LY75 and CD164
5 reference, or LY75 reference and heterozygous for a CD164 variant nucleic acid molecule, or CD164 reference and heterozygous for an LY75 variant nucleic acid molecule or heterozygous for both an LY75 variant nucleic acid molecule and a CD164 variant nucleic acid molecule, such subjects can be administered an LY75 inhibitor, a CD164 inhibitor, or both, as described herein.

For subjects that are genotyped or determined to be either or both LY75 and PARP1
10 reference, or LY75 reference and heterozygous for a PARP1 variant nucleic acid molecule, or PARP1 reference and heterozygous for an LY75 variant nucleic acid molecule or heterozygous for both an LY75 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, such subjects can be administered an LY75 inhibitor, a PARP1 inhibitor, or both, as described herein.

For subjects that are genotyped or determined to be either or both CD164 and PARP1
15 reference, or CD164 reference and heterozygous for a PARP1 variant nucleic acid molecule, or PARP1 reference and heterozygous for a CD164 variant nucleic acid molecule or heterozygous for both a CD164 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, such subjects can be administered a CD164 inhibitor, a PARP1 inhibitor, or both, as described herein.

For subjects that are genotyped or determined to be either LY75, CD164, and PARP1
20 reference; or LY75 reference and heterozygous for both a CD164 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule; or CD164 reference and heterozygous for both an LY75 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule; or PARP1 reference and heterozygous for both an LY75 variant nucleic acid molecule and a CD164 variant nucleic acid molecule; or heterozygous for an LY75 variant nucleic acid molecule, a CD164
25 variant nucleic acid molecule, and a PARP1 variant nucleic acid molecule; such subjects can be administered a LY75 inhibitor, a CD164 inhibitor, a PARP1 inhibitor or any combination thereof, as described herein.

Detecting the presence or absence of one or more LY75, CD164, or PARP1 variant nucleic acid molecules in a biological sample from a subject and/or determining whether a
30 subject has one or more LY75, CD164, or PARP1 variant nucleic acid molecules can be carried out by any of the methods described herein. In some embodiments, these methods can be carried out *in vitro*. In some embodiments, these methods can be carried out *in situ*. In some

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embodiments, these methods can be carried out *in vivo*. In any of these embodiments, the nucleic acid molecule can be present within a cell obtained from the subject.

In some embodiments, when the subject is LY75 reference, the subject is administered a therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount. In some embodiments, when the subject is heterozygous LY75 variant nucleic acid molecule, the subject is administered a therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount.

In some embodiments, when the subject is CD164 reference, the subject is administered a therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount. In some embodiments, when the subject is heterozygous for CD164 variant nucleic acid molecule, the subject is administered a therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount.

In some embodiments, when the subject is PARP1 reference, the subject is administered a therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount. In some embodiments, when the subject is heterozygous for PARP1 variant nucleic acid molecule, the subject is administered a therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount.

In some embodiments, the prevention and/or reduction methods comprise detecting the presence or absence of an LY75 predicted loss-of-function polypeptide in a biological sample from the subject. In some embodiments, when the subject does not have an LY75 predicted loss-of-function polypeptide, the subject is administered a therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount. In some embodiments, when the subject has an LY75 predicted loss-of-function polypeptide, the subject is administered a therapeutic agent that prevents or reduces CHIP in a dosage amount that is the same as or less than a standard dosage amount.

In some embodiments, the prevention and/or reduction methods comprise detecting the presence or absence of a CD164 predicted loss-of-function polypeptide in a biological sample from the subject. In some embodiments, when the subject does not have a CD164 predicted loss-of-function polypeptide, the subject is administered a therapeutic agent that

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prevents or reduces development of CHIP in a standard dosage amount. In some embodiments, when the subject has a CD164 predicted loss-of-function polypeptide, the subject is administered a therapeutic agent that prevents or reduces CHIP in a dosage amount that is the same as or less than a standard dosage amount.

5 In some embodiments, the prevention and/or reduction methods comprise detecting the presence or absence of a PARP1 predicted loss-of-function polypeptide in a biological sample from the subject. In some embodiments, when the subject does not have a PARP1 predicted loss-of-function polypeptide, the subject is administered a therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount. In some embodiments,
10 when the subject has a PARP1 predicted loss-of-function polypeptide, the subject is administered a therapeutic agent that prevents or reduces CHIP in a dosage amount that is the same as or less than a standard dosage amount.

 The present disclosure also provides methods of treating a subject with a therapeutic agent that treats or inhibits a hematologic cancer, wherein the subject has CHIP. In some
15 embodiments, the method comprises determining whether the subject has an LY75 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has an LY75 variant nucleic acid molecule. When the subject does not have an LY75 variant nucleic acid molecule, the therapeutic agent that treats or inhibits a hematologic cancer
20 is administered or continued to be administered to the subject in a standard dosage amount, and/or an LY75 inhibitor is administered to the subject. When the subject has an LY75 variant nucleic acid molecule, the therapeutic agent that treats or inhibits hematologic cancer is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor is administered to the subject.
25 The presence of an LY75 variant nucleic acid molecule indicates the subject has a decreased risk of developing hematologic cancer. In some embodiments, the subject has an LY75 variant nucleic acid molecule. In some embodiments, the subject does not have an LY75 variant nucleic acid molecule.

 The present disclosure also provides methods of treating a subject with a therapeutic
30 agent that treats or inhibits a hematologic cancer, wherein the subject has CHIP. In some embodiments, the method comprises determining whether the subject has a CD164 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and

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performing or having performed a sequence analysis on the biological sample to determine if the subject has a CD164 variant nucleic acid molecule. When the subject does not have a CD164 variant nucleic acid molecule, the therapeutic agent that treats or inhibits a hematologic cancer is administered or continued to be administered to the subject in a standard dosage amount, and/or a CD164 inhibitor is administered to the subject. When the subject has a CD164 variant nucleic acid molecule, the therapeutic agent that treats or inhibits hematologic cancer is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or a CD164 inhibitor is administered to the subject. The presence of a CD164 variant nucleic acid molecule indicates the subject has a decreased risk of developing hematologic cancer. In some embodiments, the subject has a CD164 variant nucleic acid molecule. In some embodiments, the subject does not have a CD164 variant nucleic acid molecule.

The present disclosure also provides methods of treating a subject with a therapeutic agent that treats or inhibits a hematologic cancer, wherein the subject has CHIP. In some embodiments, the method comprises determining whether the subject has a PARP1 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has a PARP1 variant nucleic acid molecule. When the subject does not have a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or inhibits a hematologic cancer is administered or continued to be administered to the subject in a standard dosage amount, and/or a PARP1 inhibitor is administered to the subject. When the subject has a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or inhibits hematologic cancer is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or a PARP1 inhibitor is administered to the subject. The presence of a PARP1 variant nucleic acid molecule indicates the subject has a decreased risk of developing hematologic cancer. In some embodiments, the subject has a PARP1 variant nucleic acid molecule. In some embodiments, the subject does not have a PARP1 variant nucleic acid molecule.

In some embodiments when the subject has both an LY75 variant nucleic acid molecule and a CD164 variant nucleic acid molecule, the therapeutic agent that treats or inhibits hematologic cancer is administered or continued to be administered to the subject in

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an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, or a CD164 inhibitor or both is administered to the subject.

In some embodiments when the subject has both an LY75 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or
5 inhibits hematologic cancer is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, or a PARP1 inhibitor, or both is administered to the subject.

In some embodiments when the subject has both a CD164 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or
10 inhibits hematologic cancer is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or a CD164 inhibitor, or a PARP1 inhibitor, or both is administered to the subject.

In some embodiments when the subject has an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and a PARP1 variant nucleic acid molecule, the
15 therapeutic agent that treats or inhibits hematologic cancer is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, a CD164 inhibitor, a PARP1 inhibitor, or any combination thereof is administered to the subject.

The present disclosure also provides methods of treating a subject with a therapeutic
20 agent that treats or inhibits an atherosclerotic cardiovascular disease, wherein the subject has CHIP. In some embodiments, the method comprises determining whether the subject has an LY75 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has an LY75 variant nucleic acid molecule. When the subject does not
25 have an LY75 variant nucleic acid molecule, the therapeutic agent that treats or inhibits an atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in a standard dosage amount, and/or an LY75 inhibitor is administered to the subject. When the subject has an LY75 variant nucleic acid molecule, the therapeutic agent that treats or inhibits atherosclerotic cardiovascular disease is administered or continued to be
30 administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor is administered to the subject. The presence of an LY75 variant nucleic acid molecule indicates the subject has a decreased risk of developing

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atherosclerotic cardiovascular disease. In some embodiments, the subject has an LY75 variant nucleic acid molecule. In some embodiments, the subject does not have an LY75 variant nucleic acid molecule.

5 The present disclosure also provides methods of treating a subject with a therapeutic agent that treats or inhibits an atherosclerotic cardiovascular disease, wherein the subject has CHIP. In some embodiments, the method comprises determining whether the subject has a CD164 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has a CD164 variant nucleic acid molecule. When the subject does
10 not have a CD164 variant nucleic acid molecule, the therapeutic agent that treats or inhibits an atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in a standard dosage amount, and/or a CD164 inhibitor is administered to the subject. When the subject has a CD164 variant nucleic acid molecule, the therapeutic agent that treats or inhibits atherosclerotic cardiovascular disease is administered or continued to be
15 administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or a CD164 inhibitor is administered to the subject. The presence of a CD164 variant nucleic acid molecule indicates the subject has a decreased risk of developing atherosclerotic cardiovascular disease. In some embodiments, the subject has a CD164 variant nucleic acid molecule. In some embodiments, the subject does not have a CD164 variant nucleic
20 acid molecule.

The present disclosure also provides methods of treating a subject with a therapeutic agent that treats or inhibits an atherosclerotic cardiovascular disease, wherein the subject has CHIP. In some embodiments, the method comprises determining whether the subject has a PARP1 variant nucleic acid molecule by obtaining or having obtained a biological sample from
25 the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has a PARP1 variant nucleic acid molecule. When the subject does not have a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or inhibits an atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in a standard dosage amount, and/or a PARP1 inhibitor is administered to the subject.
30 When the subject has a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage

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amount, and/or a PARP1 inhibitor is administered to the subject. The presence of a PARP1 variant nucleic acid molecule indicates the subject has a decreased risk of developing atherosclerotic cardiovascular disease. In some embodiments, the subject has a PARP1 variant nucleic acid molecule. In some embodiments, the subject does not have a PARP1 variant nucleic acid molecule.

In some embodiments when the subject has both an LY75 variant nucleic acid molecule and a CD164 variant nucleic acid molecule, the therapeutic agent that treats or inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, or a CD164 inhibitor or both is administered to the subject.

In some embodiments when the subject has both an LY75 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, or a PARP1 inhibitor, or both is administered to the subject.

In some embodiments when the subject has both a CD164 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or a CD164 inhibitor, or a PARP1 inhibitor, or both is administered to the subject.

In some embodiments when the subject has an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, a CD164 inhibitor, a PARP1 inhibitor, or any combination thereof is administered to the subject.

In some embodiments, the subject is heterozygous for an LY75 variant nucleic acid molecule, and the subject is further administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered the LY75 inhibitor.

In some embodiments, the subject is heterozygous for a CD164 variant nucleic acid molecule, and the subject is further administered or continued to be administered the

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therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered the CD164 inhibitor.

In some embodiments, the subject is heterozygous for a PARP1 variant nucleic acid molecule, and the subject is further administered or continued to be administered the

5 therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered the PARP1 inhibitor.

In some embodiments, the subject is heterozygous for both an LY75 variant nucleic acid molecule and a CD164 variant nucleic acid molecule, and the subject is further administered or continued to be administered the therapeutic agent that prevents or reduces
10 development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered an LY75 inhibitor, a CD164 inhibitor, or both, as described herein.

In some embodiments, the subject is heterozygous for both an LY75 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, and the subject is further administered or continued to be administered the therapeutic agent that prevents or reduces
15 development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered an LY75 inhibitor, a PARP1 inhibitor, or both, as described herein.

In some embodiments, the subject is heterozygous for both a CD164 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, and the subject is further administered or continued to be administered the therapeutic agent that prevents or reduces
20 development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered a CD164 inhibitor, a PARP1 inhibitor, or both, as described herein.

In some embodiments, the subject is heterozygous for an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and a PARP1 variant nucleic acid molecule; and the subject is further administered or continued to be administered the therapeutic agent
25 that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered an LY75 inhibitor, a CD164 inhibitor, a PARP1 inhibitor or any combination thereof, as described herein.

The present disclosure also provides methods of preventing a subject from developing CHIP by administering a therapeutic agent that prevents or reduces development of CHIP. In

30 some embodiments, the method comprises determining whether the subject has an LY75 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to

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determine if the subject has an LY75 variant nucleic acid molecule. When the subject does not have an LY75 variant nucleic acid molecule, the therapeutic agent that prevents or reduces development of CHIP is administered or continued to be administered to the subject in a standard dosage amount, and/or an LY75 inhibitor, is administered to the subject. When the
5 subject has an LY75 variant nucleic acid molecule, the therapeutic agent that prevents or reduces development of CHIP is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor is administered to the subject.

The present disclosure also provides methods of preventing a subject from developing
10 CHIP by administering a therapeutic agent that prevents or reduces development of CHIP. In some embodiments, the method comprises determining whether the subject has a CD164 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has a CD164 variant nucleic acid molecule. When the subject does not
15 have a CD164 variant nucleic acid molecule, the therapeutic agent that prevents or reduces development of CHIP is administered or continued to be administered to the subject in a standard dosage amount, and/or a CD164 inhibitor, is administered to the subject. When the subject has a CD164 variant nucleic acid molecule, the therapeutic agent that prevents or reduces development of CHIP is administered or continued to be administered to the subject in
20 an amount that is the same as or less than a standard dosage amount, and/or a CD164 inhibitor is administered to the subject.

The present disclosure also provides methods of preventing a subject from developing CHIP by administering a therapeutic agent that prevents or reduces development of CHIP. In some embodiments, the method comprises determining whether the subject has a PARP1
25 variant nucleic acid molecule by obtaining or having obtained a biological sample from the subject, and performing or having performed a sequence analysis on the biological sample to determine if the subject has a PARP1 variant nucleic acid molecule. When the subject does not have a PARP1 variant nucleic acid molecule, the therapeutic agent that prevents or reduces development of CHIP is administered or continued to be administered to the subject in a
30 standard dosage amount, and/or a PARP1 inhibitor, is administered to the subject. When the subject has a PARP1 variant nucleic acid molecule, the therapeutic agent that prevents or reduces development of CHIP is administered or continued to be administered to the subject in

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an amount that is the same as or less than a standard dosage amount, and/or a PARP1 inhibitor is administered to the subject.

In some embodiments, when the subject has both an LY75 variant nucleic acid molecule and a CD164 variant nucleic acid molecule, the therapeutic agent that treats or
5 inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, or a CD164 inhibitor or both is administered to the subject.

In some embodiments, when the subject has both an LY75 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or
10 inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, or a PARP1 inhibitor, or both is administered to the subject.

In some embodiments when the subject has both a CD164 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, the therapeutic agent that treats or
15 inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or a CD164 inhibitor, or a PARP1 inhibitor, or both is administered to the subject.

In some embodiments when the subject has an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and a PARP1 variant nucleic acid molecule, the
20 therapeutic agent that treats or inhibits atherosclerotic cardiovascular disease is administered or continued to be administered to the subject in an amount that is the same as or less than a standard dosage amount, and/or an LY75 inhibitor, a CD164 inhibitor, a PARP1 inhibitor, or any combination thereof is administered to the subject.

The presence of one or more LY75, CD164, or PARP1 variant nucleic acid molecules
25 indicates the subject has a decreased risk of developing CHIP or a CHIP-related disorder. In some embodiments, the subject has one or more LY75, CD164, or PARP1 variant nucleic acid molecules. In some embodiments, the subject does not have one or more LY75, CD164, or PARP1 variant nucleic acid molecules.

Detecting the presence or absence of one or more LY75, CD164, or PARP1 variant
30 nucleic acid molecules in a biological sample from a subject and/or determining whether a subject has one or more LY75, CD164, or PARP1 variant nucleic acid molecules can be carried out by any of the methods described herein. In some embodiments, these methods can be

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carried out *in vitro*. In some embodiments, these methods can be carried out *in situ*. In some embodiments, these methods can be carried out *in vivo*. In any of these embodiments, the polypeptide can be present within a cell obtained from the subject.

In any of the embodiments described herein for nucleic acid molecules, similar
5 methods are also included for polypeptides.

In some embodiments, the LY75 inhibitor is a small molecule. In some embodiments, the LY75 inhibitor is an inhibitory nucleic acid molecule.

In some embodiments, the CD164 inhibitor is a small molecule. In some embodiments, the CD164 inhibitor is atorvastatin. In some embodiments, the CD164 inhibitor is an inhibitory
10 nucleic acid molecule.

In some embodiments, the PARP1 inhibitor is a small molecule. In some embodiments, the PARP1 inhibitor is rucaparib, olaparib, veliparib ABT-888, veliparib, INO-1001, MK4827, CEP-9722, BMN-673, iniparib, AG-14361, NMS-P118, BYK204165, 4-hydroxyquinazoline, pamiparib, E7449, A-966492, niraparib, mortaparib, or ME0238. In some embodiments, the PARP1
15 inhibitor is an inhibitory nucleic acid molecule.

Examples of therapeutic agents that treat or inhibit myeloid neoplasia include, but are not limited to, arsenic trioxide, azacitidine, cerubidine (daunorubicin hydrochloride), cyclophosphamide, cytarabine, daunorubicin hydrochloride, daunorubicin hydrochloride and cytarabine liposome, daurismo (glasdegib maleate), dexamethasone, doxorubicin
20 hydrochloride, enasidenib mesylate, gemtuzumab ozogamicin, gilteritinib fumarate, glasdegib maleate, idamycin pfs (idarubicin hydrochloride), idarubicin hydrochloride, idhifa (enasidenib mesylate), ivosidenib, midostaurin, mitoxantrone hydrochloride, mylotarg (gemtuzumab ozogamicin), onureg (azacitidine), prednisone, rubidomycin (daunorubicin hydrochloride), rydapt (midostaurin), tabloid (thioguanine), thioguanine, tibsovo (ivosidenib), trisenox (arsenic
25 trioxide), venclexta (venetoclax), venetoclax, vincristine sulfate, vyxeos (daunorubicin hydrochloride and cytarabine liposome), and xospata (gilteritinib fumarate).

Examples of therapeutic agents that treat or inhibit lymphoid neoplasia include, but are not limited to, acalabrutinib, alemtuzumab, arzerra (ofatumumab), bendamustine hydrochloride, bendeka (bendamustine hydrochloride), calquence (acalabrutinib), campath
30 (alemtuzumab), chlorambucil, copiktra (duvelisib), cyclophosphamide, dexamethasone, duvelisib, fludarabine phosphate, gazyva (obinutuzumab), ibrutinib, idelalisib, imbruvica (ibrutinib), leukeran (chlorambucil), obinutuzumab, ofatumumab, prednisone, rituxan

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(rituximab), rituxan hycela (rituximab and hyaluronidase human), rituximab, rituximab and hyaluronidase human, treanda (bendamustine hydrochloride), truxima (rituximab), venclexta (venetoclax), venetoclax, and zydelig (idelalisib).

Examples of therapeutic agents that treat or inhibit coronary heart disease include, but are not limited to, angiotensin converting enzyme (ACE) inhibitors (such as benazepril, 5 captopril, enalapril, fosinopril, lisinopril, moexipril, perindopril, quinapril, ramipril, or trandolapril); beta blockers (such as acebutolol, atenolol, betaxolol, bisoprolol, bisoprolol/hydrochlorothiazide, metoprolol tartrate, metoprolol succinate, nadolol, pindolol, propranolol, solotol, or timolol); calcium channel blockers (such as amlodipine, diltiazem, 10 felodipine, isradipine, nifedipine, nisoldipine, or verapamil); metformin; and nitrates (such as nitroglycerin).

Examples of therapeutic agents that treat or inhibit myocardial infarction include, but are not limited to, antiplatelet blood thinners (such as aspirin, clopidogrel, prasugrel, ticagrelor, dipyridamole, or integrilin); angiotensin converting enzyme (ACE) inhibitors (such as benazepril, 15 captopril, enalapril, fosinopril, lisinopril, moexipril, perindopril, quinapril, ramipril, or trandolapril); beta blockers (such as acebutolol, atenolol, betaxolol, bisoprolol, bisoprolol/hydrochlorothiazide, metoprolol tartrate, metoprolol succinate, nadolol, pindolol, propranolol, solotol, or timolol); vasodilators (such as hydralazine or minoxidil); or thrombolytics (such as streptokinase, reteplase, alteplase, urokinase, or tenecteplase).

20 Examples of therapeutic agents that treat or inhibit severe calcified aortic valve stenosis include, but are not limited to, angiotensin converting enzyme (ACE) inhibitors (such as benazepril, captopril, enalapril, fosinopril, lisinopril, moexipril, perindopril, quinapril, ramipril, or trandolapril); beta blockers (such as acebutolol, atenolol, betaxolol, bisoprolol, bisoprolol/hydrochlorothiazide, metoprolol tartrate, metoprolol succinate, nadolol, pindolol, 25 propranolol, solotol, or timolol); diuretics (such as chlorothiazide, chlorthalidone, hydrochlorothiazide, indapamide, metolazone, bumetanide, ethacrynic acid, furosemide, torsemide, amiloride, eplerenone, spironolactone, or triamterene); and antiarrhythmic drugs (such as amiodarone, flecainide, ibutilide, lidocaine, procainamide, propafenone, quinidine, or tocainide).

30 In some embodiments, the dose of the therapeutic agents that prevent or reduce CHIP can be decreased by about 10%, by about 20%, by about 30%, by about 40%, by about 50%, by about 60%, by about 70%, by about 80%, or by about 90% for subjects that are heterozygous

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for an LY75 variant nucleic acid molecule, heterozygous for a CD164 variant nucleic acid molecule, or heterozygous for a PARP1 variant nucleic acid molecule (i.e., a less than the standard dosage amount) compared to subjects that are LY75, CD164, and PARP1 reference (who may receive a standard dosage amount). In some embodiments, the dose of the

5 therapeutic agents that prevent or reduce CHIP can be decreased by about 10%, by about 20%, by about 30%, by about 40%, or by about 50%. In addition, the subjects that are heterozygous for an LY75 variant nucleic acid molecule, or heterozygous for a CD164 variant nucleic acid molecule, or heterozygous for a PARP1 variant nucleic acid molecule can be administered less frequently compared to subjects that are LY75, CD164, and PARP1 reference.

10 In some embodiments, the dose of the therapeutic agents that prevent or reduce CHIP can be decreased by about 10%, by about 20%, by about 30%, by about 40%, by about 50%, for subjects that are homozygous for an LY75 variant nucleic acid molecule compared to subjects that are heterozygous for an LY75 variant nucleic acid molecule. In some embodiments, the dose of the therapeutic agents that prevent or reduce CHIP can be decreased by about 10%, by
15 about 20%, by about 30%, by about 40%, or by about 50%. In addition, the dose of therapeutic agents that prevent or reduce CHIP in subjects that are homozygous for an LY75 variant nucleic acid molecule can be administered less frequently compared to subjects that are heterozygous for an LY75 variant nucleic acid molecule.

In some embodiments, the dose of the therapeutic agents that prevent or reduce CHIP
20 can be decreased by about 10%, by about 20%, by about 30%, by about 40%, by about 50%, for subjects that are homozygous for a CD164 variant nucleic acid molecule compared to subjects that are heterozygous for a CD164 variant nucleic acid molecule. In some embodiments, the dose of the therapeutic agents that prevent or reduce CHIP can be decreased by about 10%, by about 20%, by about 30%, by about 40%, or by about 50%. In addition, the dose of therapeutic
25 agents that prevent or reduce CHIP in subjects that are homozygous for a CD164 variant nucleic acid molecule can be administered less frequently compared to subjects that are heterozygous for a CD164 variant nucleic acid molecule.

In some embodiments, the dose of the therapeutic agents that prevent or reduce CHIP can be decreased by about 10%, by about 20%, by about 30%, by about 40%, by about 50%, for
30 subjects that are homozygous for a PARP1 variant nucleic acid molecule compared to subjects that are heterozygous for a PARP1 variant nucleic acid molecule. In some embodiments, the dose of the therapeutic agents that prevent or reduce CHIP can be decreased by about 10%, by

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about 20%, by about 30%, by about 40%, or by about 50%. In addition, the dose of therapeutic agents that prevent or reduce CHIP in subjects that are homozygous for a PARP1 variant nucleic acid molecule can be administered less frequently compared to subjects that are heterozygous for a PARP1 variant nucleic acid molecule.

5 Administration of the therapeutic agents that prevent CHIP and/or LY75 inhibitors, or CD164 inhibitors, PARP1 inhibitors, or any combination thereof can be repeated, for example, after one day, two days, three days, five days, one week, two weeks, three weeks, one month, five weeks, six weeks, seven weeks, eight weeks, two months, or three months. The repeated administration can be at the same dose or at a different dose. The administration can be
10 repeated once, twice, three times, four times, five times, six times, seven times, eight times, nine times, ten times, or more. For example, according to certain dosage regimens a subject can receive therapy for a prolonged period of time such as, for example, 6 months, 1 year, or more.

Administration of the therapeutic agents that prevent CHIP and/or LY75 inhibitors, or
15 CD164 inhibitors, PARP1 inhibitors, or any combination thereof can occur by any suitable route including, but not limited to, parenteral, intravenous, oral, subcutaneous, intra-arterial, intracranial, intrathecal, intraperitoneal, topical, intranasal, or intramuscular. Pharmaceutical compositions for administration are desirably sterile and substantially isotonic and manufactured under GMP conditions. Pharmaceutical compositions can be provided in unit
20 dosage form (i.e., the dosage for a single administration). Pharmaceutical compositions can be formulated using one or more physiologically and pharmaceutically acceptable carriers, diluents, excipients or auxiliaries. The formulation depends on the route of administration chosen. The term "pharmaceutically acceptable" means that the carrier, diluent, excipient, or auxiliary is compatible with the other ingredients of the formulation and not substantially
25 deleterious to the recipient thereof.

The terms "treat", "treating", and "treatment" and "prevent", "preventing", and "prevention" as used herein, refer to eliciting the desired biological response, such as a therapeutic and prophylactic effect, respectively. In some embodiments, a therapeutic effect comprises one or more of a decrease/reduction in CHIP, a decrease/reduction in the severity of
30 CHIP (such as, for example, a reduction or inhibition of development of CHIP), a decrease/reduction in symptoms and CHIP-related effects, delaying the onset of symptoms and CHIP-related effects, reducing the severity of symptoms of c CHIP-related effects, reducing the

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number of symptoms and CHIP-related effects, reducing the latency of symptoms and CHIP-related effects, an amelioration of symptoms and CHIP-related effects, reducing secondary symptoms, reducing secondary infections, preventing relapse to CHIP, decreasing the number or frequency of relapse episodes, increasing latency between symptomatic episodes, increasing
5 time to sustained progression, speeding recovery, or increasing efficacy of or decreasing resistance to alternative therapeutics, and/or an increased survival time of the affected host animal, following administration of the agent or composition comprising the agent. A prophylactic effect may comprise a complete or partial avoidance/inhibition or a delay of CHIP development/progression (such as, for example, a complete or partial avoidance/inhibition or a
10 delay), and an increased survival time of the affected host animal, following administration of a therapeutic protocol. Treatment of CHIP encompasses the treatment of a subject already diagnosed as having any form of CHIP at any clinical stage or manifestation, the delay of the onset or evolution or aggravation or deterioration of the symptoms or signs of CHIP, and/or preventing and/or reducing the severity of CHIP.

15 In any of the embodiments described herein, for subjects that carry a loss-of-function variant for TET2 (such as, for example, due to the presence of an INDEL; a TET2 somatic mutation deficiency), the methods of treatment and prevention can exclude treatment with a PARP1 inhibitor. Such subjects can be otherwise treated as described herein.

The present disclosure also provides methods of identifying a subject having an
20 increased risk of developing CHIP. In some embodiments, the method comprises determining or having determined in a biological sample obtained from the subject the presence or absence of an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and/or a PARP1 variant nucleic acid molecule. When the subject lacks an LY75, a CD164, and/or a PARP1 variant nucleic acid molecule (i.e., the subject is genotypically categorized as LY75, CD164, and/or
25 PARP1 reference), then the subject has an increased risk of developing CHIP. When the subject has one or more of an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and/or a PARP1 variant nucleic acid molecule (i.e., the subject is heterozygous or homozygous for one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule), then the subject has a decreased risk of developing CHIP.

30 Having a single copy of an LY75 variant nucleic acid molecule is more protective of a subject from developing CHIP than having no copies of an LY75 variant nucleic acid molecule. Having a single copy of a CD164 variant nucleic acid molecule is more protective of a subject

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from developing CHIP than having no copies of a CD164 variant nucleic acid molecule. Having a single copy of a PARP1 variant nucleic acid molecule is more protective of a subject from developing CHIP than having no copies of a PARP1 variant nucleic acid molecule.

Without intending to be limited to any particular theory or mechanism of action, it is
5 believed that a single copy of an LY75 variant nucleic acid molecule, a CD164, variant nucleic acid molecule, or a PARP1 variant nucleic acid molecule (i.e., heterozygous for one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule) is protective of a subject from developing CHIP and CHIP-related disorders, and it is also believed that having two copies of an
10 LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, or a PARP1 variant nucleic acid molecule (i.e., homozygous for one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule) may be more protective of a subject from developing CHIP and CHIP-related disorders, relative to a subject with a single copy of a corresponding LY75, CD164, or PARP1 variant nucleic acid molecule.

Thus, in some embodiments, a single copy of of an LY75 variant nucleic acid molecule,
15 a CD164 variant nucleic acid molecule, or a PARP1 variant nucleic acid molecule may not be completely protective, but instead, may be partially or incompletely protective of a subject from developing CHIP and CHIP-related disorders. While not desiring to be bound by any particular theory, there may be additional factors or molecules involved in the development of CHIP and CHIP-related disorders that are still present in a subject having a single copy of one or
20 more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule, thus resulting in less than complete protection from the development of CHIP and CHIP-related disorders.

Determining whether a subject has one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule can be carried out by any of the methods described herein. In some embodiments, these methods can be carried out *in vitro*. In some embodiments, these
25 methods can be carried out *in situ*. In some embodiments, these methods can be carried out *in vivo*. In any of these embodiments, the nucleic acid molecule can be present within a cell obtained from the subject.

In some embodiments, when a subject is identified as having an increased risk of developing CHIP, the subject is administered a therapeutic agent that prevents or reduces
30 development of CHIP, and/or an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, as described herein. For example, when the subject is LY75 reference, and therefore has an increased risk of developing CHIP, the subject is administered an LY75

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inhibitor. In addition, when the subject is CD164 reference, and therefore has an increased risk of developing CHIP, the subject is administered a CD164 inhibitor. In addition, when the subject is PARP1 reference, and therefore has an increased risk of developing CHIP, the subject is administered a PARP1 inhibitor. In some embodiments, such a subject is also administered a
5 therapeutic agent that prevents or reduces development of CHIP.

In some embodiments, such a subject is also administered a therapeutic agent that prevents or reduces development of CHIP. In some embodiments, when the subject is homozygous for one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule, the subject is administered the therapeutic agent that prevents or reduces development of CHIP in
10 a dosage amount that is the same as or less than a standard dosage amount.

In some embodiments, when the subject is heterozygous for an LY75 variant nucleic acid molecule, the subject is administered the therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount, and is also administered an LY75 inhibitor. In some embodiments, when the subject is
15 heterozygous for a CD164 variant nucleic acid molecule, the subject is administered the therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount, and is also administered an CD164 inhibitor. In some embodiments, when the subject is heterozygous for a PARP1 variant nucleic acid molecule, the subject is administered the therapeutic agent that prevents or reduces
20 development of CHIP in a dosage amount that is the same as or less than a standard dosage amount, and is also administered an PARP1 inhibitor.

In some embodiments, the subject is heterozygous for both an LY75 variant nucleic acid molecule and a CD164 variant nucleic acid molecule, and the subject is further administered or continued to be administered the therapeutic agent that prevents or reduces
25 development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered an LY75 inhibitor, a CD164 inhibitor, or both.

In some embodiments, the subject is heterozygous for both an LY75 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, and the subject is further administered or continued to be administered the therapeutic agent that prevents or reduces
30 development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered an LY75 inhibitor, a PARP1 inhibitor, or both.

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In some embodiments, the subject is heterozygous for both a CD164 variant nucleic acid molecule and a PARP1 variant nucleic acid molecule, and the subject is further administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered a CD164 inhibitor, a PARP1 inhibitor, or both.

In some embodiments, the subject is heterozygous for an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and a PARP1 variant nucleic acid molecule, and the subject is further administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered an LY75 inhibitor, a CD164 inhibitor, a PARP1 inhibitor or any combination thereof.

In some embodiments, the subject is LY75, CD164, and PARP1 reference. In some embodiments, the subject is heterozygous for one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule. In some embodiments, the subject is homozygous for one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule.

The present disclosure also provides methods of identifying a subject having an increased risk of developing lung cancer. In some embodiments, the method comprises determining or having determined in a biological sample obtained from the subject the presence or absence of a CHIP somatic mutation in DNMT3A and/or ASXL1. When the subject lacks a CHIP somatic mutation in DNMT3A and/or ASXL1, then the subject does not have an increased risk of developing lung cancer. When the subject has a CHIP somatic mutation in DNMT3A and/or ASXL1, then the subject has an increased risk of developing lung cancer. In some embodiments, the subject is a smoker. In some embodiments, the subject is a non-smoker. In some embodiments, the CHIP somatic mutation is in DNMT3A. In some embodiments, the CHIP somatic mutation is in ASXL1. Subjects not having an increased risk of developing lung cancer because they lack a CHIP somatic mutation in DNMT3A and/or ASXL1, may still have an increased risk relative to the average individual for reasons such as smoking, breathing toxic chemicals at work, city pollution, etc.

Determining whether a subject has one or more CHIP somatic mutations in DNMT3A and/or ASXL1 can be carried out by any of the methods described herein. In some embodiments, these methods can be carried out *in vitro*. In some embodiments, these methods can be carried out *in situ*. In some embodiments, these methods can be carried out *in*

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vivo. In any of these embodiments, the nucleic acid molecule having the CHIP somatic mutation in DNMT3A and/or ASXL1 can be present within a cell obtained from the subject.

In some embodiments, when a subject is identified as having an increased risk of developing lung cancer, the subject can be subjected to enhanced monitoring, a lifestyle change, and/or lowering exposure to a hazardous substance. For example, a subject can be monitored more frequently for lung cancer pathology and/or symptoms. In some embodiments, such subjects can have chest x-rays, or the like, more frequently compared to subjects that do not have such an increased risk of developing lung cancer. In some embodiments, the subject can undergo additional monitoring for lung cancer-associated somatic mutations (such as EGFR mutations, KRAS mutations, etc.) using DNA from sputum and/or blood (i.e., more frequent cell-free DNA testing/monitoring). In some embodiments, enhanced surveillance options may include earlier magnetic resonance imaging (MRI) monitoring. In some embodiments, such subjects who are smokers can initiate smoking cessation procedures. In some embodiments, the lifestyle change can comprise lowering exposure to an environmental risk factor selected from second-hand smoke, radon, and workplace smoke exposure. In some embodiments, the hazardous substance is selected from asbestos, arsenic, nickel, chromium, beryllium, cadmium, silica, diesel exhaust, tar, or soot, or any combination thereof. In some embodiments, such subjects can undergo palliative or preventative treatment with a therapeutic agent. In some embodiments, the subject can be administered a therapeutically effective amount of erlotinib, 5-(p-methoxyphenyl)-1,2-dithiole-3-thione, deguelin, or iloprost, or any combination thereof.

In some embodiments, the subject has a CHIP somatic mutation in DNMT3A and/or ASXL1. In some embodiments, the subject does not have a CHIP somatic mutation in DNMT3A and/or ASXL1.

In any of the embodiments described herein, for subjects that are determined to have an increased risk of developing CHIP, such subjects that also carry a loss-of-function variant for TET2 (such as, for example, due to the presence of an INDEL; a TET2 somatic mutation deficiency) can undergo a treatment or prevention regimen that excludes treatment with a PARP1 inhibitor. Such subjects can be otherwise treated as described herein. Accordingly, subjects having a loss-of-function variant for TET2 and who have been determined to have an increased risk of developing CHIP can be excluded from the population of subjects amenable for treatment with a PARP1 inhibitor.

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The biological sample for detection of a CHIP somatic mutation in DNMT3A and/or ASXL1 can be derived from any cell, tissue, or biological fluid from the subject. The biological sample may comprise any clinically relevant tissue, such as lung tissue or lung cells, such as from a biopsy, a fine needle aspirate, or a sample of bodily fluid, such as blood, gingival crevicular fluid, plasma, serum, lymph, ascitic fluid, cystic fluid, or urine. In some cases, the sample comprises a buccal swab. The biological sample used in the methods disclosed herein can vary based on the assay format, nature of the detection method, and the tissues, cells, or extracts that are used as the sample. The detection of a CHIP somatic mutation in DNMT3A and/or ASXL1 can be carried out by methods similar to detection of any of the variant nucleic acid molecules described herein using the appropriate primers and probes.

The lung cancer can comprise a non-small cell lung cancer, a small cell lung cancer, mesothelioma, lung carcinoid tumor, or a chest wall tumor. In some embodiments, the lung cancer comprises a non-small cell lung cancer. In some embodiments, the lung cancer comprises a small cell lung cancer. In some embodiments, the lung cancer comprises mesothelioma. In some embodiments, the lung cancer comprises a lung carcinoid tumor. In some embodiments, the lung cancer comprises a chest wall tumor.

In some embodiments, any of the methods described herein can further comprise determining the subject's burden of having one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule, and/or one or more of an LY75, a CD164, or a PARP1 predicted loss-of-function variant polypeptide associated with a decreased risk of developing CHIP and CHIP-related disorders. The burden is the sum of all variants in the LY75 gene, the CD164 gene, and/or the PARP1 gene, which can be carried out in an association analysis with CHIP. In some embodiments, the subject is homozygous for one or more LY75 variant nucleic acid molecules associated with a decreased risk of developing CHIP. In some embodiments, the subject is heterozygous for one or more LY75 variant nucleic acid molecules associated with a decreased risk of developing CHIP. In some embodiments, the subject is homozygous for one or more CD164 variant nucleic acid molecules associated with a decreased risk of developing CHIP. In some embodiments, the subject is heterozygous for one or more CD164 variant nucleic acid molecules associated with a decreased risk of developing CHIP. In some embodiments, the subject is homozygous for one or more PARP1 variant nucleic acid molecules associated with a decreased risk of developing CHIP. In some embodiments, the subject is heterozygous for one

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or more PARP1 variant nucleic acid molecules associated with a decreased risk of developing CHIP.

The result of the association analysis suggests that LY75, CD164, and/or PARP1 variant nucleic acid molecules are associated with decreased risk of developing CHIP. When the subject has a lower burden, the subject is at a higher risk of developing CHIP and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount, and/or an LY75 inhibitor, a CD164 inhibitor, a PARP1 inhibitor, or any combination thereof. When the subject has a greater burden, the subject is at a lower risk of developing CHIP and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than the standard dosage amount. The greater the burden, the lower the risk of developing CHIP. Alternately, the gene burden analysis can comprise determining whether CHIP carriers are more likely to have any of the variants aggregated in the burden framework due to their gene effects.

In some embodiments, the subject's burden of having any one or more of an LY75, a CD164, and/or a PARP1 variant nucleic acid molecule, or an LY75 predicted loss-of-function polypeptide, a CD164 predicted loss-of-function polypeptide, and/or a PARP1 predicted loss-of-function polypeptide represents a weighted sum of a plurality of any of the LY75, CD164, or PARP1 variant nucleic acid molecules or predicted loss-of-function polypeptides. In some embodiments, the burden is calculated using at least about 2, at least about 3, at least about 4, at least about 5, at least about 10, at least about 20, at least about 30, at least about 40, at least about 50, at least about 60, at least about 70, at least about 80, at least about 100, at least about 120, at least about 150, at least about 200, at least about 250, at least about 300, at least about 400, at least about 500, at least about 1,000, at least about 10,000, at least about 100,000, or at least about or more than 1,000,000 genetic variants present in or around (up to 10 Mb) the LY75 gene, the CD164 gene, or the PARP1 gene where the genetic burden is the number of alleles multiplied by the association estimate with CHIP or related outcome for each allele (e.g., a weighted polygenic burden score). This can include any genetic variants, regardless of their genomic annotation, in proximity to any one of the LY75 gene, the CD164 gene, and/or the PARP1 gene (up to 10 Mb around the gene) that show a non-zero association with CHIP-related traits in a genetic association analysis. In some embodiments, when the subject has a burden above a desired threshold score, the subject has a decreased risk of

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developing CHIP. In some embodiments, when the subject has a burden below a desired threshold score, the subject has an increased risk of developing CHIP.

In some embodiments, the burden may be divided into quintiles, e.g., top quintile, intermediate quintile, and bottom quintile, wherein the top quintile of burden corresponds to the lowest risk group and the bottom quintile of burden corresponds to the highest risk group. In some embodiments, a subject having a greater burden comprises the highest weighted burdens, including, but not limited to the top 10%, top 20%, top 30%, top 40%, or top 50% of burdens from a subject population. In some embodiments, the genetic variants comprise the genetic variants having association with CHIP in the top 10%, top 20%, top 30%, top 40%, or top 50% of p-value range for the association. In some embodiments, each of the identified genetic variants comprise the genetic variants having association with CHIP with p-value of no more than about 10^{-2} , no more than about 10^{-3} , no more than about 10^{-4} , no more than about 10^{-5} , no more than about 10^{-6} , no more than about 10^{-7} , no more than about 10^{-8} , no more than about 10^{-9} , no more than about 10^{-10} , no more than about 10^{-11} , no more than about 10^{-12} , no more than about 10^{-13} , no more than about 10^{-14} , or no more than about or 10^{-15} . In some embodiments, the identified genetic variants comprise the genetic variants having association with CHIP with p-value of less than 5×10^{-8} . In some embodiments, the identified genetic variants comprise genetic variants having association with CHIP in high-risk subjects as compared to the rest of the reference population with odds ratio (OR) about 1.5 or greater, about 1.75 or greater, about 2.0 or greater, or about 2.25 or greater for the top 20% of the distribution; or about 1.5 or greater, about 1.75 or greater, about 2.0 or greater, about 2.25 or greater, about 2.5 or greater, or about 2.75 or greater. In some embodiments, the odds ratio (OR) may range from about 1.0 to about 1.5, from about 1.5 to about 2.0, from about 2.0 to about 2.5, from about 2.5 to about 3.0, from about 3.0 to about 3.5, from about 3.5 to about 4.0, from about 4.0 to about 4.5, from about 4.5 to about 5.0, from about 5.0 to about 5.5, from about 5.5 to about 6.0, from about 6.0 to about 6.5, from about 6.5 to about 7.0, or greater than 7.0. In some embodiments, high-risk subjects comprise subjects having burdens in the bottom decile, quintile, or tertile in a reference population. The threshold of the burden is determined on the basis of the nature of the intended practical application and the risk difference that would be considered meaningful for that practical application.

In some embodiments, when a subject is identified as having an increased risk of developing CHIP, the subject is further administered a therapeutic agent that prevents or

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reduces CHIP, and/or an LY75 inhibitor, a CD164 inhibitor, a PARP1 inhibitor, or any combination thereof, as described herein. For example, when the subject is LY75 reference, and therefore has an increased risk of developing CHIP, the subject is administered an LY75 inhibitor. In addition, when the subject CD164 reference, and therefore has an increased risk of developing CHIP, the subject is administered a CD164 inhibitor. In addition, when the subject PARP1 reference, and therefore has an increased risk of developing CHIP, the subject is administered a PARP1 inhibitor.

In some embodiments, such a subject is also administered a therapeutic agent that prevents or reduces development of CHIP. In some embodiments, when the subject is heterozygous for an LY75 variant nucleic acid molecule, the subject is administered the therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount, and is also administered an LY75 inhibitor. In some embodiments, when the subject is heterozygous for a CD164 variant nucleic acid molecule, the subject is administered the therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount, and is also administered a CD164 inhibitor. In some embodiments, when the subject is heterozygous for a PARP1 variant nucleic acid molecule, the subject is administered the therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount, and is also administered a PARP1 inhibitor.

In some embodiments, the subject is LY75, CD164, and PARP1 reference. In some embodiments, the subject is heterozygous for one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule. Furthermore, when the subject has a lower burden for having one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule, and therefore has an increased risk of developing CHIP, the subject is administered a therapeutic agent that prevents or reduces development of CHIP. In some embodiments, when the subject has a lower burden for having one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule, the subject is administered the therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or greater than the standard dosage amount administered to a subject who has a greater burden for having one or more of an LY75, a CD164, or a PARP1 variant nucleic acid molecule.

In some embodiments, any of the methods described herein can further comprise determining the subject's burden of having a CHIP somatic mutation in DNMT3A and/or ASXL1

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associated with an increased risk of developing lung cancer. The burden is the sum of all somatic mutations in the DNMT3A gene and/or ASXL1 gene, which can be carried out in an association analysis with lung cancer. In some embodiments, the subject has a CHIP somatic mutation in DNMT3A associated with an increased risk of developing lung cancer. In some

5 embodiments, the subject does not have a CHIP somatic mutation in DNMT3A associated with an increased risk of developing lung cancer. In some embodiments, the subject has a CHIP somatic mutation in ASXL1 associated with an increased risk of developing lung cancer. In some

embodiments, the subject does not have a CHIP somatic mutation in ASXL1 associated with an increased risk of developing lung cancer.

- 10 The result of the association analysis may indicate that DNMT3A and/or ASXL1 somatic mutations are associated with an increased risk of developing lung cancer. When the subject has a lower burden, the subject does not have an increased risk of developing lung cancer. When the subject has a greater burden, the subject has an increased risk of developing lung cancer, the subject can undergo any of the procedures described herein related to lung cancer.
- 15 The greater the burden, the greater the risk of developing lung cancer.

Representative DNMT3A somatic mutations include:

2:25234280:TACA CACACGCA:T	NA	frameshift	c.2727_2737delTG CGTGTGTGT	p.Phe909fs
2:25234286:C:A	NA	missense	c.2732G>T	p.Cys911Phe
2:25234286:C:T	rs906113912	missense	c.2732G>A	p.Cys911Tyr
2:25234288:CG:C		frameshift	c.2729delC	p.Ala910fs
2:25234289:G:A	rs1459396018	missense	c.2729C>T	p.Ala910Val
2:25234289:G:C	NA	missense	c.2729C>G	p.Ala910Gly
2:25234289:G:GC	NA	frameshift	c.2728dupG	p.Ala910fs
2:25234289:GC:G	NA	frameshift	c.2728delG	p.Ala910fs
2:25234290:C:CA	NA	frameshift	c.2727dupT	p.Ala910fs
2:25234290:C:G	NA	missense	c.2728G>C	p.Ala910Pro
2:25234290:C:T	NA	missense	c.2728G>A	p.Ala910Thr
2:25234290:CA:C		frameshift	c.2727delT	p.Phe909fs
2:25234290:CAA: C		frameshift	c.2726_2727delTT	p.Phe909fs
2:25234291:A:C	NA	missense	c.2727T>G	p.Phe909Leu

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2:25234291:A:T	NA	missense	c.2727T>A	p.Phe909Leu
2:25234292:A:C	NA	missense	c.2726T>G	p.Phe909Cys
2:25234292:A:G	NA	missense	c.2726T>C	p.Phe909Ser
2:25234293:A:C	NA	missense	c.2725T>G	p.Phe909Val
2:25234293:A:G	NA	missense	c.2725T>C	p.Phe909Leu
2:25234293:A:T	rs1254967511	missense	c.2725T>A	p.Phe909Ile
2:25234294:A:C	NA	stop_gained	c.2724T>G	p.Tyr908*
2:25234294:A:T	NA	stop_gained	c.2724T>A	p.Tyr908*
2:25234294:AT:A	NA	frameshift	c.2723delA	p.Tyr908fs
2:25234294:ATAC TCCT:A	NA	frameshift	c.2717_2723delAG GAGTA	p.Lys906fs
2:25234294:ATAC TCCTT:A	NA	frameshift	c.2716_2723delAA GGAGTA	p.Lys906fs
2:25234295:T:C	rs780666472	missense	c.2723A>G	p.Tyr908Cys
2:25234295:T:G	NA	missense	c.2723A>C	p.Tyr908Ser
2:25234295:T:TA	NA	frameshift	c.2722dupT	p.Tyr908fs
2:25234295:T:TA CTC	NA	frameshift	c.2719_2722dupG AGT	p.Tyr908fs
2:25234295:T:TG A	NA	frameshift	c.2722_2723insTC	p.Tyr908fs
2:25234295:TA:T	NA	frameshift	c.2722delT	p.Tyr908fs
2:25234295:TACT C:T	NA	frameshift	c.2719_2722delGA GT	p.Glu907fs
2:25234296:A:C	NA	missense	c.2722T>G	p.Tyr908Asp
2:25234296:A:T		missense	c.2722T>A	p.Tyr908Asn
2:25234296:AC:A	NA	frameshift	c.2721delG	p.Glu907fs
2:25234297:CT:C	NA	frameshift	c.2720delA	p.Glu907fs
2:25234298:T:A	NA	missense	c.2720A>T	p.Glu907Val
2:25234298:T:C	NA	missense	c.2720A>G	p.Glu907Gly
2:25234298:T:TC	NA	frameshift	c.2719dupG	p.Glu907fs
2:25234298:TC:T	NA	frameshift	c.2719delG	p.Glu907fs

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2:25234298:TCCT TCAGCGGAGCGA AG:T	NA	frameshift	c.2703_2719delCTT CGCTCCGCTGAAGG	p.Phe902fs
2:25234299:C:A	NA	stop_gained	c.2719G>T	p.Glu907*
2:25234300:C:A	NA	missense	c.2718G>T	p.Lys906Asn
2:25234300:C:CT	NA	frameshift	c.2717dupA	p.Glu907fs
2:25234300:C:G	NA	missense	c.2718G>C	p.Lys906Asn
2:25234300:CT:C	NA	frameshift	c.2717delA	p.Lys906fs
2:25234300:CTTC A:C	NA	frameshift	c.2714_2717delTG AA	p.Leu905fs
2:25234302:T:A	NA	stop_gained	c.2716A>T	p.Lys906*
2:25234302:T:C	rs1240890824	missense	c.2716A>G	p.Lys906Glu
2:25234302:TCAG CGGAGCGAAGAG GTGG:T	NA	frameshift	c.2697_2715delCC ACCTCTTCGCTCCG CTG	p.His900fs
2:25234303:CA:C	NA	frameshift	c.2714delT	p.Leu905fs
2:25234303:CAGC GG:C	NA	frameshift	c.2710_2714delCC GCT	p.Pro904fs
2:25234303:CAGC GGAGCGA:C	NA	frameshift	c.2705_2714delTC GCTCCGCT	p.Phe902fs
2:25234303:CAGC GGAGCGAAG:C	NA	inframe_indel	c.2703_2714delCTT CGCTCCGCT	p.Phe902_Leu905 del
2:25234304:A:C	rs751868166	missense	c.2714T>G	p.Leu905Arg
2:25234304:A:G		missense	c.2714T>C	p.Leu905Pro
2:25234304:A:T	rs751868166	missense	c.2714T>A	p.Leu905Gln
2:25234304:AGC GGAGCGAAGAG: A	NA	frameshift	c.2701_2713delCT CTTCGCTCCGC	p.Leu901fs
2:25234305:G:C	NA	missense	c.2713C>G	p.Leu905Val
2:25234305:G:T	NA	missense	c.2713C>A	p.Leu905Met
2:25234306:C:CG	NA	frameshift	c.2711dupC	p.Leu905fs
2:25234306:CG:C	NA	frameshift	c.2711delC	p.Pro904fs
2:25234306:CGG AGCGAAGAG:C	NA	frameshift	c.2701_2711delCT CTTCGCTCC	p.Leu901fs

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2:25234307:G:A	rs149095705	missense	c.2711C>T	p.Pro904Leu
2:25234307:G:C	rs149095705	missense	c.2711C>G	p.Pro904Arg
2:25234307:G:GG AGC	NA	frameshift	c.2707_2710dupGC TC	p.Pro904fs
2:25234307:G:T	rs149095705	missense	c.2711C>A	p.Pro904Gln
2:25234307:GGA GC:G	NA	frameshift	c.2707_2710delGC TC	p.Ala903fs
2:25234307:GGA GCGAA:G	NA	frameshift	c.2704_2710delTTC GCTC	p.Phe902fs
2:25234308:G:A	NA	missense	c.2710C>T	p.Pro904Ser
2:25234308:G:C	NA	missense	c.2710C>G	p.Pro904Ala
2:25234308:GAG CGA:G	NA	frameshift	c.2705_2709delTC GCT	p.Phe902fs
2:25234309:AGC: A	NA	frameshift	c.2707_2708delGC	p.Ala903fs
2:25234309:AGC GAAGAGGTG:A	NA	frameshift	c.2698_2708delCA CCTCTTCGC	p.His900fs
2:25234310:G:A	NA	missense	c.2708C>T	p.Ala903Val
2:25234310:G:T	NA	missense	c.2708C>A	p.Ala903Asp
2:25234310:GC:G	rs1323968409	frameshift	c.2707delG	p.Ala903fs
2:25234311:C:CG	NA	frameshift	c.2706dupC	p.Ala903fs
2:25234311:C:CG AAGAGGTGGCGG ATGACTGGCACGC TCCATGACCGGCC CA	NA	frameshift	c.2666_2706dupTG GGCCGGTCATGGA GCGTGCCAGTCATC CGCCACCTCTTC	p.Ala903fs
2:25234311:C:G	NA	missense	c.2707G>C	p.Ala903Pro
2:25234311:C:T	rs749167103	missense	c.2707G>A	p.Ala903Thr
2:25234311:CG:C		frameshift	c.2706delC	p.Phe902fs
2:25234312:G:GA	NA	frameshift	c.2705dupT	p.Ala903fs
2:25234312:G:GT	NA	frameshift	c.2705_2706insA	p.Phe902fs
2:25234312:GAA: G	NA	frameshift	c.2704_2705delTT	p.Phe902fs

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2:25234313:A:AA GAGG	NA	frameshift	c.2700_2704dupCC TCT	p.Phe902fs
2:25234313:A:G	rs587777510	missense	c.2705T>C	p.Phe902Ser
2:25234313:A:T	NA	missense	c.2705T>A	p.Phe902Tyr
2:25234314:A:C	NA	missense	c.2704T>G	p.Phe902Val
2:25234315:G:GA	NA	frameshift	c.2702dupT	p.Phe902fs
2:25234315:GA:G	NA	frameshift	c.2702delT	p.Leu901fs
2:25234316:A:C	rs774384800	missense	c.2702T>G	p.Leu901Arg
2:25234316:A:G	rs774384800	missense	c.2702T>C	p.Leu901Pro
2:25234316:A:T	NA	missense	c.2702T>A	p.Leu901His
2:25234316:AG:A	NA	frameshift	c.2701delC	p.Leu901fs
2:25234317:G:A	NA	missense	c.2701C>T	p.Leu901Phe
2:25234317:G:C	rs1360815999	missense	c.2701C>G	p.Leu901Val
2:25234318:GT:G	NA	frameshift	c.2699delA	p.His900fs
2:25234319:T:A	NA	missense	c.2699A>T	p.His900Leu
2:25234319:T:G	NA	missense	c.2699A>C	p.His900Pro
2:25234319:TG:T	NA	frameshift	c.2698delC	p.His900fs
2:25234320:G:A	NA	missense	c.2698C>T	p.His900Tyr
2:25234320:G:GG CGGATGA	NA	frameshift	c.2690_2697dupTC ATCCGC	p.His900fs
2:25234322:C:A	NA	missense	c.2696G>T	p.Arg899Leu
2:25234322:C:G	rs1480791034	missense	c.2696G>C	p.Arg899Pro
2:25234322:C:T	NA	missense	c.2696G>A	p.Arg899His
2:25234322:CG:C	NA	frameshift	c.2695delC	p.Arg899fs
2:25234323:G:A	rs771922296	missense	c.2695C>T	p.Arg899Cys
2:25234323:G:C	NA	missense	c.2695C>G	p.Arg899Gly
2:25234323:G:T	NA	missense	c.2695C>A	p.Arg899Ser
2:25234324:G:GA TGACTGGCACGCT CC	NA	frameshift	c.2678_2693dupG GAGCGTGCCAGTCA T	p.Ile898fs
2:25234324:GA:G	NA	frameshift	c.2693delT	p.Ile898fs

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2:25234324:GAT GACTGGCACGCTC C:G	NA	frameshift	c.2678_2693delGG AGCGTGCCAGTCAT	p.Trp893fs
2:25234325:A:AT	NA	frameshift	c.2692dupA	p.Ile898fs
2:25234325:A:C	NA	missense	c.2693T>G	p.Ile898Ser
2:25234325:A:G	NA	missense	c.2693T>C	p.Ile898Thr
2:25234325:A:T	rs960019580	missense	c.2693T>A	p.Ile898Asn
2:25234326:T:C	NA	missense	c.2692A>G	p.Ile898Val
2:25234327:GA:G	NA	frameshift	c.2690delT	p.Val897fs
2:25234327:GACT GGC:G	NA	inframe_indel	c.2685_2690delGC CAGT	p.Pro896_Val897 del
2:25234328:A:C	NA	missense	c.2690T>G	p.Val897Gly
2:25234328:A:T	NA	missense	c.2690T>A	p.Val897Asp
2:25234328:ACTG G:A	NA	frameshift	c.2686_2689delCC AG	p.Pro896fs
2:25234329:C:CT	NA	frameshift	c.2688dupA	p.Val897fs
2:25234329:C:CT GGCACGGTCA	NA	frameshift	c.2688_2689insTG ACCGTGCCA	p.Val897fs
2:25234329:C:G	NA	missense	c.2689G>C	p.Val897Leu
2:25234329:CT:C	NA	frameshift	c.2688delA	p.Val897fs
2:25234330:T:TG GCACGCTCCATGA CC	NA	frameshift	c.2672_2687dupG GTCATGGAGCGTGC C	p.Ile898fs
2:25234330:T:TG GGC	NA	frameshift	c.2687_2688insGC CC	p.Val897fs
2:25234330:TG:T	NA	frameshift	c.2687delC	p.Pro896fs
2:25234331:G:A	NA	missense	c.2687C>T	p.Pro896Leu
2:25234332:G:GA ACGCTC	NA	frameshift	c.2685_2686insGA GCGTT	p.Pro896fs
2:25234333:C:CA	NA	frameshift	c.2684dupT	p.Pro896fs
2:25234333:C:CA CGCT	NA	frameshift	c.2680_2684dupA GCGT	p.Pro896fs
2:25234334:A:T	NA	missense	c.2684T>A	p.Val895Glu
2:25234335:C:T	rs762126968	missense	c.2683G>A	p.Val895Met

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2:25234336:G:C	NA	missense	c.2682C>G	p.Ser894Arg
2:25234336:GCTC CATGACCGGCCCA GCAGTCT:G	NA	frameshift	c.2659_2681delAG ACTGCTGGGCCGGT CATGGAG	p.Leu888fs
2:25234337:C:CT	NA	frameshift	c.2680dupA	p.Ser894fs
2:25234337:C:T	NA	missense	c.2681G>A	p.Ser894Asn
2:25234338:T:C	NA	missense	c.2680A>G	p.Ser894Gly
2:25234338:T:TC	NA	frameshift	c.2679dupG	p.Ser894fs
2:25234338:T:TCC ATGACCGGCCC	NA	frameshift	c.2667_2679dupG GGCCGGTCATGG	p.Ser894fs
2:25234339:C:A	NA	missense	c.2679G>T	p.Trp893Cys
2:25234339:C:T	rs1330526945	stop_gained	c.2679G>A	p.Trp893*
2:25234340:C:G	NA	missense	c.2678G>C	p.Trp893Ser
2:25234340:C:T	rs750515748	stop_gained	c.2678G>A	p.Trp893*
2:25234341:A:G	NA	missense	c.2677T>C	p.Trp893Arg
2:25234341:A:T	NA	missense	c.2677T>A	p.Trp893Arg
2:25234342:T:TG	NA	frameshift	c.2675dupC	p.Trp893fs
2:25234342:TGAC C:T	NA	frameshift	c.2672_2675delGG TC	p.Arg891fs
2:25234343:G:A	rs1355266109	missense	c.2675C>T	p.Ser892Leu
2:25234343:G:C	NA	stop_gained	c.2675C>G	p.Ser892*
2:25234343:G:T	NA	stop_gained	c.2675C>A	p.Ser892*
2:25234343:GA:G	NA	frameshift	c.2674delT	p.Ser892fs
2:25234344:A:AC CGG	NA	frameshift	c.2670_2673dupCC GG	p.Ser892fs
2:25234344:AC:A	NA	frameshift	c.2673delG	p.Ser892fs
2:25234344:ACCG G:A	NA	frameshift	c.2670_2673delCC GG	p.Arg891fs
2:25234344:ACCG GCCCAGCAGTC:A	NA	frameshift	c.2660_2673delGA CTGCTGGGCCGG	p.Arg887fs
2:25234346:C:T	rs1389510486	missense	c.2672G>A	p.Arg891Gln
2:25234346:CG:C	rs762357545	frameshift	c.2671delC	p.Arg891fs
2:25234347:G:A	NA	missense	c.2671C>T	p.Arg891Trp

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2:25234347:G:C	NA	missense	c.2671C>G	p.Arg891Gly
2:25234347:G:GG CCC	NA	frameshift	c.2667_2670dupG GGC	p.Arg891fs
2:25234348:G:GC	rs766070325	frameshift	c.2669dupG	p.Arg891fs
2:25234348:G:GC C	NA	frameshift	c.2668_2669dupG G	p.Arg891fs
2:25234348:GC:G	NA	frameshift	c.2669delG	p.Gly890fs
2:25234348:GCC: G	NA	frameshift	c.2668_2669delGG	p.Gly890fs
2:25234349:C:T	rs752160822	missense	c.2669G>A	p.Gly890Asp
2:25234349:CCCA GCAGTCT:C	NA	frameshift	c.2659_2668delAG ACTGCTGG	p.Arg887fs
2:25234350:C:G	NA	missense	c.2668G>C	p.Gly890Arg
2:25234350:C:T	NA	missense	c.2668G>A	p.Gly890Ser
2:25234351:CAGC AGTCTCT:C	NA	frameshift	c.2657_2666delAG AGACTGCT	p.Gln886fs
2:25234352:A:C	NA	missense	c.2666T>G	p.Leu889Arg
2:25234352:A:G	NA	missense	c.2666T>C	p.Leu889Pro
2:25234352:A:T	NA	missense	c.2666T>A	p.Leu889Gln
2:25234354:C:CA G	NA	frameshift	c.2662_2663dupCT	p.Leu889fs
2:25234354:CA:C	NA	frameshift	c.2663delT	p.Leu888fs
2:25234354:CAG: C	NA	frameshift	c.2662_2663delCT	p.Leu888fs
2:25234355:A:G	NA	missense	c.2663T>C	p.Leu888Pro
2:25234355:A:T	NA	missense	c.2663T>A	p.Leu888Gln
2:25234355:AG:A	NA	frameshift	c.2662delC	p.Leu888fs
2:25234355:AGTC TCTGCCTCGCCAA GCG:A	NA	frameshift	c.2644_2662delCG CTTGGCGAGGCAG AGAC	p.Arg882fs
2:25234356:G:GT C	NA	frameshift	c.2660_2661dupG A	p.Leu888fs
2:25234356:GT:G	NA	frameshift	c.2661delA	p.Arg887fs

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2:25234356:GTC: G	NA	frameshift	c.2660_2661delGA	p.Arg887fs
2:25234356:GTCT C:G	NA	frameshift	c.2658_2661delGA GA	p.Gln886fs
2:25234357:T:G	NA	missense	c.2661A>C	p.Arg887Ser
2:25234357:TCTC TGC:T	NA	inframe_indel	c.2655_2660delGC AGAG	p.Gln886_Arg887 del
2:25234358:C:A	NA	missense	c.2660G>T	p.Arg887Ile
2:25234359:T:A	NA	stop_gained	c.2659A>T	p.Arg887*
2:25234359:T:C	NA	missense	c.2659A>G	p.Arg887Gly
2:25234360:C:CG	NA	frameshift	c.2657_2658insC	p.Gln886fs
2:25234360:CT:C	NA	frameshift	c.2657delA	p.Gln886fs
2:25234361:T:C	NA	missense	c.2657A>G	p.Gln886Arg
2:25234362:G:A	rs752280049	stop_gained	c.2656C>T	p.Gln886*
2:25234362:G:C	rs752280049	missense	c.2656C>G	p.Gln886Glu
2:25234362:G:T	NA	missense	c.2656C>A	p.Gln886Lys
2:25234362:GC:G	NA	frameshift	c.2655delG	p.Arg885fs
2:25234364:C:A	NA	missense	c.2654G>T	p.Arg885Met
2:25234364:C:T	NA	missense	c.2654G>A	p.Arg885Lys
2:25234364:CT:C	NA	frameshift	c.2653delA	p.Arg885fs
2:25234365:TC:T	NA	frameshift	c.2652delG	p.Arg885fs
2:25234366:CGCC AAGCGGCTCAT:C	NA	frameshift	c.2638_2651delAT GAGCCGCTTGGC	p.Met880fs
2:25234367:G:A	rs559023562	missense	c.2651C>T	p.Ala884Val
2:25234367:G:GC C	NA	frameshift	c.2649_2650dupG G	p.Ala884fs
2:25234367:GC:G		frameshift	c.2650delG	p.Ala884fs
2:25234367:GCCA AGCGGCT:G	NA	frameshift	c.2641_2650delAG CCGCTTGG	p.Ser881fs
2:25234369:C:A	NA	missense	c.2649G>T	p.Leu883Phe
2:25234369:CA:C	NA	frameshift	c.2648delT	p.Leu883fs
2:25234370:A:AC	NA	frameshift	c.2647_2648insG	p.Leu883fs
2:25234370:A:T	rs1307673132	stop_gained	c.2648T>A	p.Leu883*

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2:25234372:GCG GCTCATGTTGGAG A:G	NA	frameshift	c.2630_2645delTCT CCAACATGAGCCG	p.Val877fs
2:25234373:C:A	rs147001633	missense	c.2645G>T	p.Arg882Leu
2:25234373:C:G	rs147001633	missense	c.2645G>C	p.Arg882Pro
2:25234373:C:T	rs147001633	missense	c.2645G>A	p.Arg882His
2:25234373:CG:C	NA	frameshift	c.2644delC	p.Arg882fs
2:25234373:CGG CTCATGTTG:C	NA	frameshift	c.2634_2644delCA ACATGAGCC	p.Asn879fs
2:25234374:G:A	rs377577594	missense	c.2644C>T	p.Arg882Cys
2:25234374:G:C	rs377577594	missense	c.2644C>G	p.Arg882Gly
2:25234374:G:T	rs377577594	missense	c.2644C>A	p.Arg882Ser
2:25234376:C:CT CATGTTGGAGAC G	NA	frameshift	c.2628_2641dupCG TCTCCAACATGA	p.Ser881fs
2:25234376:CT:C	NA	frameshift	c.2641delA	p.Ser881fs
2:25234377:T:TC A	NA	frameshift	c.2639_2640dupTG	p.Ser881fs
2:25234378:C:A	NA	missense	c.2640G>T	p.Met880Ile
2:25234378:C:CA	NA	frameshift	c.2639dupT	p.Met880fs
2:25234378:C:G	NA	missense	c.2640G>C	p.Met880Ile
2:25234378:C:T	NA	missense	c.2640G>A	p.Met880Ile
2:25234378:CA:C	NA	frameshift	c.2639delT	p.Met880fs
2:25234379:AT:A	NA	frameshift	c.2638delA	p.Met880fs
2:25234379:ATGT T:A	NA	frameshift	c.2635_2638delAA CA	p.Asn879fs
2:25234379:ATGT TGGAGACGTCAG TATAGTGGAC:A	NA	frameshift	c.2614_2638delGT CCACTATACTGACG TCTCCAACA	p.Val872fs
2:25234380:T:A	NA	missense	c.2638A>T	p.Met880Leu
2:25234380:T:C	rs775365749	missense	c.2638A>G	p.Met880Val
2:25234380:TG:T	NA	frameshift	c.2637delC	p.Asn879fs
2:25234381:GT:G	NA	frameshift	c.2636delA	p.Asn879fs
2:25234382:T:C	NA	missense	c.2636A>G	p.Asn879Ser

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2:25234383:T:C	rs1188120438	missense	c.2635A>G	p.Asn879Asp
2:25234383:T:TG	NA	frameshift	c.2634dupC	p.Asn879fs
2:25234383:T:TG G	NA	frameshift	c.2633_2634dupCC	p.Asn879fs
2:25234384:G:GG A	NA	frameshift	c.2632_2633dupTC	p.Asn879fs
2:25234385:GAG ACGTC:G	NA	frameshift	c.2626_2632delGA CGTCT	p.Asp876fs
2:25234386:A:G	NA	missense	c.2632T>C	p.Ser878Pro
2:25234387:GA:G	NA	frameshift	c.2630delT	p.Val877fs
2:25234388:AC:A	NA	frameshift	c.2629delG	p.Val877fs
2:25234389:C:A	NA	missense	c.2629G>T	p.Val877Phe
2:25234389:C:CG TCA	NA	frameshift	c.2625_2628dupTG AC	p.Val877fs
2:25234389:C:T	NA	missense	c.2629G>A	p.Val877Ile
2:25234389:CG:C	NA	frameshift	c.2628delC	p.Asp876fs
2:25234389:CGTC A:C	NA	frameshift	c.2625_2628delTG AC	p.Asp876fs
2:25234390:G:GT	rs774080362	frameshift	c.2627dupA	p.Asp876fs
2:25234390:G:T	NA	missense	c.2628C>A	p.Asp876Glu
2:25234390:GT:G	NA	frameshift	c.2627delA	p.Asp876fs
2:25234391:T:A	NA	missense	c.2627A>T	p.Asp876Val
2:25234391:T:C	NA	missense	c.2627A>G	p.Asp876Gly
2:25234391:T:G	NA	missense	c.2627A>C	p.Asp876Ala
2:25234391:T:TC	NA	frameshift	c.2626dupG	p.Asp876fs
2:25234391:TC:T	NA	frameshift	c.2626delG	p.Asp876fs
2:25234391:TCA: T	NA	frameshift	c.2625_2626delTG	p.Asp876fs
2:25234392:C:G	NA	missense	c.2626G>C	p.Asp876His
2:25234392:C:T	rs992291948	missense	c.2626G>A	p.Asp876Asn
2:25234392:CA:C	NA	frameshift	c.2625delT	p.Asp876fs
2:25234392:CAGT AT:C	NA	frameshift	c.2621_2625delAT ACT	p.Tyr874fs

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2:25234394:G:A	NA	missense	c.2624C>T	p.Thr875Ile
2:25234394:G:C	NA	missense	c.2624C>G	p.Thr875Ser
2:25234394:G:GT A	NA	frameshift	c.2622_2623dupTA	p.Thr875fs
2:25234394:GTA: G	NA	frameshift	c.2622_2623delTA	p.Thr875fs
2:25234395:T:C	NA	missense	c.2623A>G	p.Thr875Ala
2:25234396:A:AT	NA	frameshift	c.2621dupA	p.Tyr874fs
2:25234396:A:C	NA	stop_gained	c.2622T>G	p.Tyr874*
2:25234396:A:T	rs770134132	stop_gained	c.2622T>A	p.Tyr874*
2:25234397:T:A	NA	missense	c.2621A>T	p.Tyr874Phe
2:25234397:T:C	NA	missense	c.2621A>G	p.Tyr874Cys
2:25234397:TA:T	NA	frameshift	c.2620delT	p.Tyr874fs
2:25234398:A:T	NA	missense	c.2620T>A	p.Tyr874Asn
2:25234399:G:C	NA	missense	c.2619C>G	p.His873Gln
2:25234400:T:C	rs1280811819	missense	c.2618A>G	p.His873Arg
2:25234400:T:TG	NA	frameshift	c.2617dupC	p.His873fs
2:25234400:TG:T	NA	frameshift	c.2617delC	p.His873fs
2:25234400:TGG ACTGG:T	NA	frameshift	c.2611_2617delCC AGTCC	p.Pro871fs
2:25234401:G:A	rs773610723	missense	c.2617C>T	p.His873Tyr
2:25234401:G:C	NA	missense	c.2617C>G	p.His873Asp
2:25234403:AC:A	NA	frameshift	c.2614delG	p.Val872fs
2:25234403:ACTG GGAAACCAAATAC C:A	NA	frameshift	c.2598_2614delGG TATTTGGTTTCCCA G	p.Arg866fs
2:25234404:C:T	NA	missense	c.2614G>A	p.Val872Ile
2:25234404:CT:C	NA	frameshift	c.2613delA	p.Val872fs
2:25234405:T:TG G	NA	frameshift	c.2611_2612dupCC	p.Val872fs
2:25234405:TG:T	rs760010938	frameshift	c.2612delC	p.Pro871fs
2:25234405:TGG: T	NA	frameshift	c.2611_2612delCC	p.Pro871fs

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2:25234406:G:A	rs1419990763	missense	c.2612C>T	p.Pro871Leu
2:25234407:G:C	NA	missense	c.2611C>G	p.Pro871Ala
2:25234408:GA:G	NA	frameshift	c.2609delT	p.Phe870fs
2:25234408:GAA: G	NA	frameshift	c.2608_2609delTT	p.Phe870fs
2:25234409:A:C	NA	missense	c.2609T>G	p.Phe870Cys
2:25234411:AC:A	NA	frameshift	c.2606delG	p.Gly869fs
2:25234412:C:A	NA	missense	c.2606G>T	p.Gly869Val
2:25234412:C:G	NA	missense	c.2606G>C	p.Gly869Ala
2:25234412:C:T	NA	missense	c.2606G>A	p.Gly869Asp
2:25234413:C:CA AAT	NA	frameshift	c.2601_2604dupAT TT	p.Gly869fs
2:25234413:C:G	NA	missense	c.2605G>C	p.Gly869Arg
2:25234413:C:T		missense	c.2605G>A	p.Gly869Ser
2:25234413:CA:C	rs753089697	frameshift	c.2604delT	p.Phe868fs
2:25234414:A:C	rs1239050589	missense	c.2604T>G	p.Phe868Leu
2:25234414:A:T	NA	missense	c.2604T>A	p.Phe868Leu
2:25234415:A:C	NA	missense	c.2603T>G	p.Phe868Cys
2:25234415:A:G	NA	missense	c.2603T>C	p.Phe868Ser
2:25234415:A:T	NA	missense	c.2603T>A	p.Phe868Tyr
2:25234416:A:C	NA	missense	c.2602T>G	p.Phe868Val
2:25234416:A:G	NA	missense	c.2602T>C	p.Phe868Leu
2:25234417:T:TA	NA	frameshift	c.2600dupT	p.Phe868fs
2:25234417:TACC CTGGGGGAGAGAA AGGCAGAGAGGG CA:T	NA	splice_acceptor	c.2598- 26_2600delTGCCCT CTCTGCCTTTTCTCC CCCAGGGT	p.Val867del
2:25234418:AC:A	rs761283545	frameshift	c.2599delG	p.Val867fs
2:25234418:ACCC TGGGG:A	NA	frameshift	c.2598- 6_2599delCCCCAG GG	p.Arg866fs
2:25234420:CCTG :C	NA	splice_acceptor	c.2598-3_2598- 1delCAG	

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2:25234420:CCTG GGG:C	NA	splice_acceptor	c.2598-6_2598-1delCCCCAG	
2:25234421:C:A	rs766506181	splice_acceptor	c.2598-1G>T	
2:25234421:C:G	rs766506181	splice_acceptor	c.2598-1G>C	
2:25234421:C:T	rs766506181	splice_acceptor	c.2598-1G>A	
2:25234421:CT:C	NA	splice_acceptor	c.2598-2delA	
2:25234422:T:A	NA	splice_acceptor	c.2598-2A>T	
2:25234422:T:C	rs760092360	splice_acceptor	c.2598-2A>G	
2:25234422:T:G	NA	splice_acceptor	c.2598-2A>C	
2:25235702:GGT ACCTTTCCATTTC :G	NA	splice_donor	c.2586_2597+4delT GAAATGGAAAGGT AC	p.Glu863_Arg866 del
2:25235705:A:C	NA	splice_donor	c.2597+2T>G	
2:25235705:A:G	NA	splice_donor	c.2597+2T>C	
2:25235705:AC:A	NA	splice_donor	c.2597+1delG	
2:25235706:C:T	rs764855628	splice_donor	c.2597+1G>A	
2:25235707:C:CT	NA	frameshift	c.2596dupA	p.Arg866fs
2:25235707:C:CTT TCCATTTCAGTGC ACCATAAG	NA	frameshift	c.2574_2596dupCT TATGGTGCACTGAA ATGGAAA	p.Arg866fs
2:25235707:C:T	NA	missense	c.2597G>A	p.Arg866Lys
2:25235707:CT:C	NA	frameshift	c.2596delA	p.Arg866fs
2:25235710:T:A	rs1187041748	missense	c.2594A>T	p.Glu865Val
2:25235710:T:C	NA	missense	c.2594A>G	p.Glu865Gly
2:25235710:T:G	NA	missense	c.2594A>C	p.Glu865Ala
2:25235711:C:CC ATTT	NA	frameshift	c.2588_2592dupAA ATG	p.Glu865fs

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2:25235712:CA:C	NA	frameshift	c.2591delT	p.Met864fs
2:25235713:A:G	NA	missense	c.2591T>C	p.Met864Thr
2:25235713:AT:A	NA	frameshift	c.2590delA	p.Met864fs
2:25235713:ATTT CAGTGCACCATAA GATGTCCTC:A	NA	frameshift	c.2566_2590delGA GGACATCTTATGGT GCACTGAAA	p.Glu856fs
2:25235716:T:A	NA	missense	c.2588A>T	p.Glu863Val
2:25235717:C:A	NA	stop_gained	c.2587G>T	p.Glu863*
2:25235717:C:T	NA	missense	c.2587G>A	p.Glu863Lys
2:25235718:A:AG T	NA	frameshift	c.2584_2585dupAC	p.Glu863fs
2:25235718:AGT: A	NA	frameshift	c.2584_2585delAC	p.Thr862fs
2:25235718:AGT GCACCAT:A	NA	inframe_indel	c.2577_2585delAT GGTGCAC	p.Leu859_Thr862 delinsPhe
2:25235719:G:A	NA	missense	c.2585C>T	p.Thr862Ile
2:25235719:G:T	NA	missense	c.2585C>A	p.Thr862Asn
2:25235719:GTGC ACCA:G	NA	frameshift	c.2578_2584delTG GTGCA	p.Trp860fs
2:25235720:TG:T	NA	frameshift	c.2583delC	p.Cys861fs
2:25235721:G:T	NA	stop_gained	c.2583C>A	p.Cys861*
2:25235722:C:T	NA	missense	c.2582G>A	p.Cys861Tyr
2:25235722:CACC A:C	NA	frameshift	c.2578_2581delTG GT	p.Trp860fs
2:25235723:A:AC	NA	frameshift	c.2580dupG	p.Cys861fs
2:25235723:A:G	NA	missense	c.2581T>C	p.Cys861Arg
2:25235723:ACCA TAAGATGTCCTCT TTCTCATTC:A	NA	frameshift	c.2556_2580delGA ATGAGAAAGAGGA CATCTTATGG	p.Met852fs
2:25235724:C:G	NA	missense	c.2580G>C	p.Trp860Cys
2:25235724:C:T	rs376830288	stop_gained	c.2580G>A	p.Trp860*
2:25235725:C:G	NA	missense	c.2579G>C	p.Trp860Ser
2:25235725:C:T	rs1321116253	stop_gained	c.2579G>A	p.Trp860*

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2:25235725:CAT: C	NA	frameshift	c.2577_2578delAT	p.Trp860fs
2:25235725:CATA A:C	NA	frameshift	c.2575_2578delTT AT	p.Leu859fs
2:25235726:A:AT	rs764502627	frameshift	c.2577dupA	p.Trp860fs
2:25235726:A:C	NA	missense	c.2578T>G	p.Trp860Gly
2:25235726:A:G	rs373014701	missense	c.2578T>C	p.Trp860Arg
2:25235726:A:T	NA	missense	c.2578T>A	p.Trp860Arg
2:25235726:ATAA GATG:A	NA	frameshift	c.2571_2577delCA TCTTA	p.Ile858fs
2:25235726:ATAA GATGTCCTCT:A	NA	frameshift	c.2565_2577delAG AGGACATCTTA	p.Lys855fs
2:25235726:ATAA GATGTCCTCTTTC: A	NA	frameshift	c.2562_2577delGA AAGAGGACATCTTA	p.Glu854fs
2:25235727:T:TC	NA	frameshift	c.2576_2577insG	p.Trp860fs
2:25235727:TA:T	NA	frameshift	c.2576delT	p.Leu859fs
2:25235728:A:C	rs1175622747	stop_gained	c.2576T>G	p.Leu859*
2:25235728:A:T	NA	stop_gained	c.2576T>A	p.Leu859*
2:25235732:TG:T	NA	frameshift	c.2571delC	p.Asp857fs
2:25235734:T:TCC	NA	frameshift	c.2568_2569dupG G	p.Asp857fs
2:25235734:TC:T	rs1254668110	frameshift	c.2569delG	p.Asp857fs
2:25235735:C:T	NA	missense	c.2569G>A	p.Asp857Asn
2:25235735:CCT: C	rs754004583	frameshift	c.2567_2568delAG	p.Glu856fs
2:25235736:CTCT T:C	NA	frameshift	c.2564_2567delAA GA	p.Lys855fs
2:25235737:TCTT TCTCATTCATG:T	NA	frameshift	c.2553_2566delCA TGAATGAGAAAG	p.Phe851fs
2:25235738:C:A	NA	stop_gained	c.2566G>T	p.Glu856*
2:25235738:C:CT	NA	frameshift	c.2565dupA	p.Glu856fs
2:25235738:CT:C	NA	frameshift	c.2565delA	p.Glu856fs
2:25235738:CTT:C	NA	frameshift	c.2564_2565delAA	p.Lys855fs

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2:25235740:T:A	NA	missense	c.2564A>T	p.Lys855Ile
2:25235740:TTCT C:T	NA	frameshift	c.2560_2563delGA GA	p.Glu854fs
2:25235741:T:A	NA	stop_gained	c.2563A>T	p.Lys855*
2:25235742:CTCA TT:C	NA	frameshift	c.2557_2561delAA TGA	p.Asn853fs
2:25235743:TC:T	NA	frameshift	c.2560delG	p.Glu854fs
2:25235744:CATT CATG:C	NA	frameshift	c.2553_2559delCA TGAAT	p.Phe851fs
2:25235745:AT:A	NA	frameshift	c.2558delA	p.Asn853fs
2:25235748:C:CA	NA	frameshift	c.2555dupT	p.Met852fs
2:25235749:A:T	NA	missense	c.2555T>A	p.Met852Lys
2:25235749:ATG: A	NA	frameshift	c.2553_2554delCA	p.Met852fs
2:25235749:ATGA AGACAGGAAAAT GC:A	NA	frameshift	c.2538_2554delGC ATTTTCCTGTCTTCA	p.Gln846fs
2:25235750:T:C	rs746967478	missense	c.2554A>G	p.Met852Val
2:25235750:T:G	NA	missense	c.2554A>C	p.Met852Leu
2:25235750:T:TG	NA	frameshift	c.2553dupC	p.Met852fs
2:25235750:TG:T	rs757223824	frameshift	c.2553delC	p.Phe851fs
2:25235751:G:GA	NA	frameshift	c.2552dupT	p.Met852fs
2:25235751:GA:G	NA	frameshift	c.2552delT	p.Phe851fs
2:25235751:GAA GA:G	NA	frameshift	c.2549_2552delTCT T	p.Val850fs
2:25235751:GAA GAC:G	NA	frameshift	c.2548_2552delGT CTT	p.Val850fs
2:25235752:AAG: A	rs779108270	frameshift	c.2550_2551delCT	p.Phe851fs
2:25235754:G:GA	NA	frameshift	c.2549dupT	p.Phe851fs
2:25235754:GAC: G	NA	frameshift	c.2548_2549delGT	p.Val850fs
2:25235755:AC:A	rs1175091304	frameshift	c.2548delG	p.Val850fs
2:25235756:C:G	NA	missense	c.2548G>C	p.Val850Leu

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2:25235756:C:T	NA	missense	c.2548G>A	p.Val850Ile
2:25235756:CA:C	NA	frameshift	c.2547delT	p.Val850fs
2:25235756:CAG: C	NA	frameshift	c.2546_2547delCT	p.Pro849fs
2:25235757:AG:A	rs750600685	frameshift	c.2546delC	p.Pro849fs
2:25235758:G:A	NA	missense	c.2546C>T	p.Pro849Leu
2:25235758:G:C	NA	missense	c.2546C>G	p.Pro849Arg
2:25235758:G:GG A	NA	frameshift	c.2544_2545dupTC	p.Pro849fs
2:25235758:G:T	NA	missense	c.2546C>A	p.Pro849His
2:25235759:G:A	NA	missense	c.2545C>T	p.Pro849Ser
2:25235759:G:C	NA	missense	c.2545C>G	p.Pro849Ala
2:25235759:G:GA	NA	frameshift	c.2544dupT	p.Pro849fs
2:25235759:G:T	NA	missense	c.2545C>A	p.Pro849Thr
2:25235759:GA:G	NA	frameshift	c.2544delT	p.Pro849fs
2:25235762:A:C	NA	missense	c.2542T>G	p.Phe848Val
2:25235762:A:T	NA	missense	c.2542T>A	p.Phe848Ile
2:25235763:AT:A	NA	frameshift	c.2540delA	p.His847fs
2:25235764:TG:T	NA	frameshift	c.2539delC	p.His847fs
2:25235764:TGC: T	NA	frameshift	c.2538_2539delGC	p.His847fs
2:25235764:TGCT GG:T	NA	frameshift	c.2535_2539delCC AGC	p.Asp845fs
2:25235765:G:T	NA	missense	c.2539C>A	p.His847Asn
2:25235765:GC:G	NA	frameshift	c.2538delG	p.Gln846fs
2:25235766:CT:C	NA	frameshift	c.2537delA	p.Gln846fs
2:25235766:CTG: C	NA	frameshift	c.2536_2537delCA	p.Gln846fs
2:25235767:TG:T	NA	frameshift	c.2536delC	p.Gln846fs
2:25235768:G:A	rs781139634	stop_gained	c.2536C>T	p.Gln846*
2:25235769:G:C	NA	missense	c.2535C>G	p.Asp845Glu
2:25235770:TC:T	NA	frameshift	c.2533delG	p.Asp845fs
2:25235771:CTT:C	NA	frameshift	c.2531_2532delAA	p.Lys844fs

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2:25235771:CTTT GCCCTGCTTT:C	NA	frameshift	c.2520_2532delAA AGCAGGGCAAA	p.Ile840fs
2:25235774:T:C	NA	missense	c.2530A>G	p.Lys844Glu
2:25235774:TG:T	NA	frameshift	c.2529delC	p.Asp845fs
2:25235775:GC:G	NA	frameshift	c.2528delG	p.Gly843fs
2:25235776:C:CC CTG	NA	frameshift	c.2524_2527dupCA GG	p.Gly843fs
2:25235776:C:T	NA	missense	c.2528G>A	p.Gly843Asp
2:25235777:C:G	NA	missense	c.2527G>C	p.Gly843Arg
2:25235778:C:G	NA	missense	c.2526G>C	p.Gln842His
2:25235778:CT:C	NA	frameshift	c.2525delA	p.Gln842fs
2:25235779:T:C	rs771174392	missense	c.2525A>G	p.Gln842Arg
2:25235779:TG:T	NA	frameshift	c.2524delC	p.Gln842fs
2:25235779:TGCT TTATG:T	NA	frameshift	c.2517_2524delCA TAAAGC	p.Ile840fs
2:25235780:G:A	rs1015295548	stop_gained	c.2524C>T	p.Gln842*
2:25235780:G:C	NA	missense	c.2524C>G	p.Gln842Glu
2:25235780:GC:G	NA	frameshift	c.2523delG	p.Lys841fs
2:25235781:C:CTT TATGGAGTT	NA	frameshift	c.2512_2522dupAA CTCCATAAA	p.Gln842fs
2:25235781:CT:C	NA	frameshift	c.2522delA	p.Lys841fs
2:25235781:CTT:C	NA	frameshift	c.2521_2522delAA	p.Lys841fs
2:25235783:T:C	rs1404111595	missense	c.2521A>G	p.Lys841Glu
2:25235783:T:G	NA	missense	c.2521A>C	p.Lys841Gln
2:25235783:TTA:T	NA	frameshift	c.2519_2520delTA	p.Ile840fs
2:25235784:TA:T	NA	frameshift	c.2519delT	p.Ile840fs
2:25235786:T:C	NA	missense	c.2518A>G	p.Ile840Val
2:25235786:T:TG	NA	frameshift	c.2517dupC	p.Ile840fs
2:25235786:T:TG G	NA	frameshift	c.2516_2517dupCC	p.Ile840fs
2:25235786:TG:T	NA	frameshift	c.2517delC	p.Ile840fs
2:25235787:G:GA	NA	frameshift	c.2516_2517insT	p.Ile840fs
2:25235788:G:A	NA	missense	c.2516C>T	p.Ser839Phe

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2:25235788:GA:G	NA	frameshift	c.2515delT	p.Ser839fs
2:25235789:A:C	NA	missense	c.2515T>G	p.Ser839Ala
2:25235789:AG:A	NA	frameshift	c.2514delC	p.Ser839fs
2:25235790:G:GT	rs781008582	frameshift	c.2513dupA	p.Asn838fs
2:25235790:GT:G	NA	frameshift	c.2513delA	p.Asn838fs
2:25235792:T:C	rs961377711	missense	c.2512A>G	p.Asn838Asp
2:25235794:G:C	rs1374267987	stop_gained	c.2510C>G	p.Ser837*
2:25235794:G:GA TCA	NA	frameshift	c.2509_2510insTG AT	p.Ser837fs
2:25235794:G:T	NA	stop_gained	c.2510C>A	p.Ser837*
2:25235795:A:AC	NA	frameshift	c.2508dupG	p.Ser837fs
2:25235797:C:G	NA	missense	c.2507G>C	p.Arg836Thr
2:25235800:G:A	rs1489843853	missense	c.2504C>T	p.Thr835Met
2:25235800:GT:G	rs1293842202	frameshift	c.2503delA	p.Thr835fs
2:25235801:T:A	NA	missense	c.2503A>T	p.Thr835Ser
2:25235803:G:A	NA	missense	c.2501C>T	p.Thr834Ile
2:25235803:G:GT	NA	frameshift	c.2500dupA	p.Thr834fs
2:25235804:T:C	NA	missense	c.2500A>G	p.Thr834Ala
2:25235804:T:TA ATGGTCCTCACTT TGCTG	NA	frameshift	c.2481_2499dupCA GCAAAGTGAGGAC CATT	p.Thr834fs
2:25235804:TA:T	NA	frameshift	c.2499delT	p.Thr834fs
2:25235806:A:T	NA	missense	c.2498T>A	p.Ile833Asn
2:25235807:T:C	rs745931882	missense	c.2497A>G	p.Ile833Val
2:25235809:GT:G	NA	frameshift	c.2494delA	p.Thr832fs
2:25235812:C:T	NA	missense	c.2492G>A	p.Arg831Lys
2:25235820:GCTG AACTAGATGAAG A:G	NA	frameshift	c.2479- 11_2483delTCTTCA TCTAGTTCAG	p.Phe827fs
2:25235821:CTGA ACT:C	NA	frameshift	c.2479- 2_2482delAGTTCA	p.Phe827fs
2:25235822:T:TG AACTAGAG	NA	stop_gained	c.2481_2482insCTC TAGTTC	p.Phe827_Ser828i nsLeuTerPhe

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2:25235822:TG:T	rs769831202	frameshift	c.2481delC	p.Phe827fs
2:25235823:G:GT	NA	frameshift	c.2480_2481insA	p.Phe827fs
2:25235823:G:T	NA	missense	c.2481C>A	p.Phe827Leu
2:25235823:GA:G	NA	frameshift	c.2480delT	p.Phe827fs
2:25235823:GAA: G	NA	frameshift	c.2479_2480delTT	p.Phe827fs
2:25235825:A:C	NA	missense	c.2479T>G	p.Phe827Val
2:25235826:C:A	NA	splice_acceptor	c.2479-1G>T	
2:25235826:C:G	NA	splice_acceptor	c.2479-1G>C	
2:25235826:C:T	rs775933506	splice_acceptor	c.2479-1G>A	
2:25235826:CT:C	NA	splice_acceptor	c.2479-2delA	
2:25235827:T:C	rs761171028	splice_acceptor	c.2479-2A>G	
2:25235827:T:G	NA	splice_acceptor	c.2479-2A>C	
2:25236934:A:AC	NA	splice_donor	c.2478+1dupG	
2:25236934:A:C	rs1327946633	splice_donor	c.2478+2T>G	
2:25236934:A:G	NA	splice_donor	c.2478+2T>C	
2:25236934:A:T	NA	splice_donor	c.2478+2T>A	
2:25236934:AC:A	NA	splice_donor	c.2478+1delG	
2:25236935:C:A	NA	splice_donor	c.2478+1G>T	
2:25236935:C:G	NA	splice_donor	c.2478+1G>C	
2:25236935:C:T	rs762213449	splice_donor	c.2478+1G>A	
2:25236935:CCT: C	NA	frameshift	c.2477_2478delAG	p.Lys826fs
2:25236936:C:G	NA	missense	c.2478G>C	p.Lys826Asn
2:25236936:CT:C	NA	frameshift	c.2477delA	p.Lys826fs
2:25236936:CTTG :C	NA	inframe_indel	c.2475_2477delCA A	p.Lys826del
2:25236937:T:A	NA	missense	c.2477A>T	p.Lys826Met

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2:25236937:T:C	rs770079872	missense	c.2477A>G	p.Lys826Arg
2:25236937:T:G	rs770079872	missense	c.2477A>C	p.Lys826Thr
2:25236938:T:C	NA	missense	c.2476A>G	p.Lys826Glu
2:25236939:GGCT ATCC:G	NA	frameshift	c.2468_2474delGG ATAGC	p.Arg823fs
2:25236940:G:T	NA	missense	c.2474C>A	p.Ala825Asp
2:25236941:C:T	NA	missense	c.2473G>A	p.Ala825Thr
2:25236942:TA:T	NA	frameshift	c.2471delT	p.Ile824fs
2:25236943:A:G	NA	missense	c.2471T>C	p.Ile824Thr
2:25236943:AT:A	NA	frameshift	c.2470delA	p.Ile824fs
2:25236944:T:TC	rs1200243711	frameshift	c.2469dupG	p.Ile824fs
2:25236944:TC:T	NA	frameshift	c.2469delG	p.Ile824fs
2:25236946:C:T	NA	missense	c.2468G>A	p.Arg823Lys
2:25236946:CT:C	NA	frameshift	c.2467delA	p.Arg823fs
2:25236947:T:C	NA	missense	c.2467A>G	p.Arg823Gly
2:25236948:G:GC	NA	frameshift	c.2465dupG	p.Arg823fs
2:25236948:GC:G	NA	frameshift	c.2465delG	p.Gly822fs
2:25236950:CA:C	NA	frameshift	c.2463delT	p.His821fs
2:25236951:A:C	NA	missense	c.2463T>G	p.His821Gln
2:25236951:A:T	NA	missense	c.2463T>A	p.His821Gln
2:25236951:AT:A	NA	frameshift	c.2462delA	p.His821fs
2:25236952:T:A	NA	missense	c.2462A>T	p.His821Leu
2:25236952:T:C	NA	missense	c.2462A>G	p.His821Arg
2:25236952:T:G	rs772882388	missense	c.2462A>C	p.His821Pro
2:25236952:TG:T	NA	frameshift	c.2461delC	p.His821fs
2:25236955:T:A	NA	missense	c.2459A>T	p.Glu820Val
2:25236955:T:C	NA	missense	c.2459A>G	p.Glu820Gly
2:25236955:TC:T	NA	frameshift	c.2458delG	p.Glu820fs
2:25236955:TCCA G:T	NA	frameshift	c.2455_2458delCT GG	p.Leu819fs
2:25236956:C:A	rs1264543822	stop_gained	c.2458G>T	p.Glu820*
2:25236956:C:G	NA	missense	c.2458G>C	p.Glu820Gln

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2:25236956:CCA: C	NA	frameshift	c.2456_2457delTG	p.Leu819fs
2:25236956:CCAG A:C	NA	frameshift	c.2454_2457delTCT G	p.Cys818fs
2:25236957:CA:C	NA	frameshift	c.2456delT	p.Leu819fs
2:25236958:A:T	NA	missense	c.2456T>A	p.Leu819Gln
2:25236958:AG:A	NA	frameshift	c.2455delC	p.Leu819fs
2:25236959:G:C	NA	missense	c.2455C>G	p.Leu819Val
2:25236959:G:T	NA	missense	c.2455C>A	p.Leu819Met
2:25236960:A:AC ACT	NA	frameshift	c.2450_2453dupA GTG	p.Cys818fs
2:25236960:A:AC C	NA	frameshift	c.2453_2454insGG	p.Cys818fs
2:25236960:A:T	NA	stop_gained	c.2454T>A	p.Cys818*
2:25236960:AC:A	NA	frameshift	c.2453delG	p.Cys818fs
2:25236961:C:A	NA	missense	c.2453G>T	p.Cys818Phe
2:25236961:C:T	NA	missense	c.2453G>A	p.Cys818Tyr
2:25236961:CA:C	NA	frameshift	c.2452delT	p.Cys818fs
2:25236962:A:C	NA	missense	c.2452T>G	p.Cys818Gly
2:25236962:A:G	NA	missense	c.2452T>C	p.Cys818Arg
2:25236962:A:T	NA	missense	c.2452T>A	p.Cys818Ser
2:25236963:C:CT	NA	frameshift	c.2450dupA	p.Cys818fs
2:25236963:CT:C	NA	frameshift	c.2450delA	p.Glu817fs
2:25236964:T:C	NA	missense	c.2450A>G	p.Glu817Gly
2:25236964:TC:T	NA	frameshift	c.2449delG	p.Glu817fs
2:25236965:C:A	rs373873045	stop_gained	c.2449G>T	p.Glu817*
2:25236966:C:CT GCA	NA	frameshift	c.2444_2447dupTG CA	p.Gln816fs
2:25236966:CT:C	NA	frameshift	c.2447delA	p.Gln816fs
2:25236967:T:C	NA	missense	c.2447A>G	p.Gln816Arg
2:25236967:TG:T	NA	frameshift	c.2446delC	p.Gln816fs
2:25236968:G:A	rs759187608	stop_gained	c.2446C>T	p.Gln816*
2:25236969:C:CA	NA	frameshift	c.2444dupT	p.Gln816fs

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2:25236969:CA:C	NA	frameshift	c.2444delT	p.Leu815fs
2:25236970:A:G	NA	missense	c.2444T>C	p.Leu815Pro
2:25236970:A:T	NA	missense	c.2444T>A	p.Leu815Gln
2:25236970:AG:A	NA	frameshift	c.2443delC	p.Leu815fs
2:25236971:G:C	NA	missense	c.2443C>G	p.Leu815Val
2:25236971:G:T	NA	missense	c.2443C>A	p.Leu815Met
2:25236972:CT:C	NA	frameshift	c.2441delA	p.Glu814fs
2:25236972:CTCC AGCT:C	NA	frameshift	c.2435_2441delAG CTGGA	p.Lys812fs
2:25236973:T:C	NA	missense	c.2441A>G	p.Glu814Gly
2:25236973:T:TC	NA	frameshift	c.2440dupG	p.Glu814fs
2:25236974:C:A	rs1174921909	stop_gained	c.2440G>T	p.Glu814*
2:25236975:CA:C	NA	frameshift	c.2438delT	p.Leu813fs
2:25236977:G:GC TTATCA	NA	frameshift	c.2430_2436dupTG ATAAG	p.Leu813fs
2:25236977:GC:G	NA	frameshift	c.2436delG	p.Lys812fs
2:25236977:GCTT AT:G	NA	frameshift	c.2432_2436delAT AAG	p.Asp811fs
2:25236978:CT:C	NA	frameshift	c.2435delA	p.Lys812fs
2:25236979:T:C	NA	missense	c.2435A>G	p.Lys812Arg
2:25236979:T:TTA	NA	frameshift	c.2433_2434dupTA	p.Lys812fs
2:25236980:T:A	NA	stop_gained	c.2434A>T	p.Lys812*
2:25236980:T:TA	rs1287022880	frameshift	c.2433dupT	p.Lys812fs
2:25236980:TATC ATTCACAGTGG:T	NA	frameshift	c.2420_2433delCC ACTGTGAATGAT	p.Ser807fs
2:25236981:A:AT CAT	NA	frameshift	c.2429_2432dupAT GA	p.Asp811fs
2:25236981:A:T	NA	missense	c.2433T>A	p.Asp811Glu
2:25236981:ATC: A	NA	frameshift	c.2431_2432delGA	p.Asp811fs
2:25236981:ATCA TT:A	NA	frameshift	c.2428_2432delAA TGA	p.Asn810fs
2:25236983:CA:C	NA	frameshift	c.2430delT	p.Asn810fs

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2:25236984:AT:A	NA	frameshift	c.2429delA	p.Asn810fs
2:25236986:TCA: T	NA	frameshift	c.2426_2427delTG	p.Val809fs
2:25236989:CA:C	NA	frameshift	c.2424delT	p.Val809fs
2:25236990:AG:A	NA	frameshift	c.2423delC	p.Thr808fs
2:25236991:GTG GA:G	NA	frameshift	c.2419_2422delTC CA	p.Ser807fs
2:25236991:GTG GATGCCAACGGCC :G	NA	frameshift	c.2409- 1_2422delGGCCGT TGGCATCCA	p.Arg803fs
2:25236994:G:GA TGC	NA	frameshift	c.2416_2419dupGC AT	p.Ser807fs
2:25236994:G:T	NA	missense	c.2420C>A	p.Ser807Tyr
2:25236997:GC:G	NA	frameshift	c.2416delG	p.Ala806fs
2:25236998:C:T	NA	missense	c.2416G>A	p.Ala806Thr
2:25236999:C:CA	NA	frameshift	c.2414dupT	p.Leu805fs
2:25236999:C:CT	NA	frameshift	c.2414_2415insA	p.Ala806fs
2:25236999:CA:C	NA	frameshift	c.2414delT	p.Leu805fs
2:25237000:A:AA AAT	NA	frameshift	c.2413_2414insATT T	p.Leu805fs
2:25237000:A:T	NA	stop_gained	c.2414T>A	p.Leu805*
2:25237002:CG:C	NA	frameshift	c.2411delC	p.Pro804fs
2:25237002:CGG CCTAGGAGGCAG AAGA:C	NA	splice_acceptor	c.2409- 15_2411delTCTTCT GCCTCCTAGGCC	p.Pro804del
2:25237003:G:A	rs750597155	missense	c.2411C>T	p.Pro804Leu
2:25237003:G:T	NA	missense	c.2411C>A	p.Pro804Gln
2:25237003:GGC: G	NA	frameshift	c.2409_2410delGC	p.Arg803fs
2:25237004:G:A	NA	missense	c.2410C>T	p.Pro804Ser
2:25237004:GC:G	rs35824014	frameshift	c.2409delG	p.Arg803fs
2:25237005:C:A	rs1304201075	missense	c.2409G>T	p.Arg803Ser
2:25237005:C:G	NA	missense	c.2409G>C	p.Arg803Ser

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2:25237006:C:A	rs866917013	splice_acceptor	c.2409-1G>T	
2:25237006:C:G	rs866917013	splice_acceptor	c.2409-1G>C	
2:25237006:C:T	rs866917013	splice_acceptor	c.2409-1G>A	
2:25237007:T:A	NA	splice_acceptor	c.2409-2A>T	
2:25237007:T:C	NA	splice_acceptor	c.2409-2A>G	
2:25237007:T:G	NA	splice_acceptor	c.2409-2A>C	
2:25239128:A:C	NA	splice_donor	c.2408+2T>G	
2:25239128:A:G	NA	splice_donor	c.2408+2T>C	
2:25239128:A:T	NA	splice_donor	c.2408+2T>A	
2:25239129:C:A	NA	splice_donor	c.2408+1G>T	
2:25239129:C:G	NA	splice_donor	c.2408+1G>C	
2:25239129:C:T	rs1457044151	splice_donor	c.2408+1G>A	
2:25239130:C:A	NA	missense	c.2408G>T	p.Arg803Met
2:25239130:C:G	NA	missense	c.2408G>C	p.Arg803Thr
2:25239130:C:T	rs764146514	missense	c.2408G>A	p.Arg803Lys
2:25239131:T:A	NA	missense	c.2407A>T	p.Arg803Trp
2:25239131:T:C	NA	missense	c.2407A>G	p.Arg803Gly
2:25239131:TG:T	NA	frameshift	c.2406delC	p.Asn802fs
2:25239131:TGTT CATACCGGGAA:T	NA	frameshift	c.2393_2406delTTC CCGGTATGAAC	p.Leu798fs
2:25239132:G:GA	NA	frameshift	c.2405_2406insT	p.Arg803fs
2:25239132:GT:G	NA	frameshift	c.2405delA	p.Asn802fs
2:25239135:C:A	NA	missense	c.2403G>T	p.Met801Ile
2:25239135:C:T	NA	missense	c.2403G>A	p.Met801Ile
2:25239136:A:C	rs901395842	missense	c.2402T>G	p.Met801Arg
2:25239136:A:G	NA	missense	c.2402T>C	p.Met801Thr
2:25239136:A:T	NA	missense	c.2402T>A	p.Met801Lys

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2:25239136:AT:A	NA	frameshift	c.2401delA	p.Met801fs
2:25239136:ATAC CGGGAAGG:A	NA	frameshift	c.2391_2401delCC TTCCCGGTA	p.Leu798fs
2:25239137:T:A	NA	missense	c.2401A>T	p.Met801Leu
2:25239137:T:C	rs753567076	missense	c.2401A>G	p.Met801Val
2:25239138:AC:A	NA	frameshift	c.2399delG	p.Gly800fs
2:25239139:C:A	NA	missense	c.2399G>T	p.Gly800Val
2:25239139:C:T	NA	missense	c.2399G>A	p.Gly800Asp
2:25239140:C:CG	NA	frameshift	c.2397dupC	p.Gly800fs
2:25239140:C:T	rs757083492	missense	c.2398G>A	p.Gly800Ser
2:25239140:CG:C	rs998597777	frameshift	c.2397delC	p.Gly800fs
2:25239142:G:A	rs1004794515	missense	c.2396C>T	p.Pro799Leu
2:25239142:G:C	rs1004794515	missense	c.2396C>G	p.Pro799Arg
2:25239143:G:A	rs766564570	missense	c.2395C>T	p.Pro799Ser
2:25239143:G:C	rs766564570	missense	c.2395C>G	p.Pro799Ala
2:25239143:G:T	rs766564570	missense	c.2395C>A	p.Pro799Thr
2:25239144:AAG GT:A	NA	frameshift	c.2390_2393delAC CT	p.Asn797fs
2:25239144:AAG GTTACCCC:A	NA	frameshift	c.2384_2393delGG GGTAACCT	p.Trp795fs
2:25239144:AAG GTTACCCCAG:A	NA	inframe_indel	c.2382_2393delCT GGGGTAACCT	p.Trp795_Leu798 del
2:25239145:A:C	NA	missense	c.2393T>G	p.Leu798Arg
2:25239145:A:G	rs1399762821	missense	c.2393T>C	p.Leu798Pro
2:25239145:A:T	NA	missense	c.2393T>A	p.Leu798His
2:25239145:AG:A	NA	frameshift	c.2392delC	p.Leu798fs
2:25239145:AGG TTACCC:A	NA	frameshift	c.2385_2392delGG GTAACC	p.Trp795fs
2:25239146:G:A	NA	missense	c.2392C>T	p.Leu798Phe
2:25239146:G:C	NA	missense	c.2392C>G	p.Leu798Val
2:25239146:G:GG TAA	NA	frameshift	c.2391_2392insTTA C	p.Gly800fs

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2:25239146:G:GG TAAC	NA	frameshift	c.2391_2392insGTT AC	p.Leu798fs
2:25239147:G:C	NA	missense	c.2391C>G	p.Asn797Lys
2:25239147:G:GC	NA	frameshift	c.2390_2391insG	p.Asn797fs
2:25239147:G:T		missense	c.2391C>A	p.Asn797Lys
2:25239147:GT:G	rs748836168	frameshift	c.2390delA	p.Asn797fs
2:25239148:T:A	NA	missense	c.2390A>T	p.Asn797Ile
2:25239148:T:C	rs767588813	missense	c.2390A>G	p.Asn797Ser
2:25239148:T:G	NA	missense	c.2390A>C	p.Asn797Thr
2:25239148:TTAC CCCA:T	NA	frameshift	c.2383_2389delTG GGGTA	p.Trp795fs
2:25239149:T:A	NA	missense	c.2389A>T	p.Asn797Tyr
2:25239149:T:C	rs755013083	missense	c.2389A>G	p.Asn797Asp
2:25239149:T:G	rs755013083	missense	c.2389A>C	p.Asn797His
2:25239149:TACC CCAGA:T	NA	frameshift	c.2381_2388delTCT GGGGT	p.Phe794fs
2:25239150:A:AC C	NA	frameshift	c.2386_2387dupG G	p.Asn797fs
2:25239150:AC:A	rs1025684646	frameshift	c.2387delG	p.Gly796fs
2:25239150:ACCC CAG:A	NA	inframe_indel	c.2382_2387delCT GGGG	p.Trp795_Gly796 del
2:25239151:C:A	rs781138077	missense	c.2387G>T	p.Gly796Val
2:25239151:C:G	NA	missense	c.2387G>C	p.Gly796Ala
2:25239151:C:T	rs781138077	missense	c.2387G>A	p.Gly796Asp
2:25239152:C:A	NA	missense	c.2386G>T	p.Gly796Cys
2:25239152:C:G	NA	missense	c.2386G>C	p.Gly796Arg
2:25239153:C:A	NA	missense	c.2385G>T	p.Trp795Cys
2:25239153:C:G	NA	missense	c.2385G>C	p.Trp795Cys
2:25239153:C:T	rs1395575712	stop_gained	c.2385G>A	p.Trp795*
2:25239154:C:A	rs756566100	missense	c.2384G>T	p.Trp795Leu
2:25239154:C:G	NA	missense	c.2384G>C	p.Trp795Ser
2:25239154:C:T	rs756566100	stop_gained	c.2384G>A	p.Trp795*
2:25239154:CA:C	NA	frameshift	c.2383delT	p.Trp795fs

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2:25239154:CAG: C	NA	frameshift	c.2382_2383delCT	p.Phe794fs
2:25239154:CAG A:C	NA	inframe_indel	c.2381_2383delTCT	p.Phe794del
2:25239155:A:G	rs778414705	missense	c.2383T>C	p.Trp795Arg
2:25239155:A:T		missense	c.2383T>A	p.Trp795Arg
2:25239156:G:C	NA	missense	c.2382C>G	p.Phe794Leu
2:25239157:A:C	NA	missense	c.2381T>G	p.Phe794Cys
2:25239157:A:G	rs1419173604	missense	c.2381T>C	p.Phe794Ser
2:25239158:A:C		missense	c.2380T>G	p.Phe794Val
2:25239158:A:G	rs771474959	missense	c.2380T>C	p.Phe794Leu
2:25239158:A:T	NA	missense	c.2380T>A	p.Phe794Ile
2:25239159:G:C	rs1418024882	stop_gained	c.2379C>G	p.Tyr793*
2:25239159:G:T	NA	stop_gained	c.2379C>A	p.Tyr793*
2:25239160:T:G	NA	missense	c.2378A>C	p.Tyr793Ser
2:25239161:A:C	rs1247759759	missense	c.2377T>G	p.Tyr793Asp
2:25239161:A:G	NA	missense	c.2377T>C	p.Tyr793His
2:25239163:C:G	NA	missense	c.2375G>C	p.Arg792Pro
2:25239163:C:T	rs774100557	missense	c.2375G>A	p.Arg792His
2:25239163:CG:C	NA	frameshift	c.2374delC	p.Arg792fs
2:25239164:G:A	NA	missense	c.2374C>T	p.Arg792Cys
2:25239164:G:C	NA	missense	c.2374C>G	p.Arg792Gly
2:25239164:G:GG GCCCTGT	NA	frameshift	c.2366_2373dupAC AGGGCC	p.Arg792fs
2:25239164:GGG CCCTGTGT:G	NA	frameshift	c.2364_2373delAC ACAGGGCC	p.His789fs
2:25239166:G:A	NA	missense	c.2372C>T	p.Ala791Val
2:25239166:G:T	NA	missense	c.2372C>A	p.Ala791Asp
2:25239166:GC:G	NA	frameshift	c.2371delG	p.Ala791fs
2:25239167:C:T	rs1005623505	missense	c.2371G>A	p.Ala791Thr
2:25239167:CCCT GT:C	NA	frameshift	c.2366_2370delAC AGG	p.His789fs
2:25239168:C:G	NA	missense	c.2370G>C	p.Arg790Ser

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2:25239169:CT:C	NA	frameshift	c.2368delA	p.Arg790fs
2:25239169:CTG: C	NA	frameshift	c.2367_2368delCA	p.His789fs
2:25239170:T:A	NA	missense	c.2368A>T	p.Arg790Trp
2:25239171:G:T	NA	missense	c.2367C>A	p.His789Gln
2:25239172:T:A	NA	missense	c.2366A>T	p.His789Leu
2:25239173:GT:G	NA	frameshift	c.2364delA	p.His789fs
2:25239175:G:A	rs1277979168	missense	c.2363C>T	p.Ala788Val
2:25239175:GCA GCT:G	NA	frameshift	c.2358_2362delAG CTG	p.Ala787fs
2:25239176:C:T	NA	missense	c.2362G>A	p.Ala788Thr
2:25239176:CA:C	NA	frameshift	c.2361delT	p.Ala788fs
2:25239177:AGC: A	NA	frameshift	c.2359_2360delGC	p.Ala787fs
2:25239178:G:GC	NA	frameshift	c.2359dupG	p.Ala787fs
2:25239178:GCT: G	NA	frameshift	c.2358_2359delAG	p.Ala787fs
2:25239179:C:A	rs375845138	missense	c.2359G>T	p.Ala787Ser
2:25239179:C:CT	NA	frameshift	c.2358dupA	p.Ala787fs
2:25239179:C:T	NA	missense	c.2359G>A	p.Ala787Thr
2:25239181:G:A	NA	missense	c.2357C>T	p.Ser786Leu
2:25239181:G:C	rs962973415	stop_gained	c.2357C>G	p.Ser786*
2:25239181:G:T	NA	stop_gained	c.2357C>A	p.Ser786*
2:25239181:GAC: G	NA	frameshift	c.2355_2356delGT	p.Ala787fs
2:25239185:C:T	NA	missense	c.2353G>A	p.Val785Met
2:25239186:TTC:T	NA	frameshift	c.2350_2351delGA	p.Glu784fs
2:25239187:T:TC AAA	NA	frameshift	c.2350_2351insTTT G	p.Glu784fs
2:25239187:T:TCT TTGGCATCAATCA TCACAGGGTTGG A	NA	frameshift	c.2323_2350dupTC CAACCCTGTGATGA TTGATGCCAAAG	p.Glu784fs
2:25239187:TC:T	NA	frameshift	c.2350delG	p.Glu784fs

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2:25239188:C:A	NA	stop_gained	c.2350G>T	p.Glu784*
2:25239188:C:CT	NA	frameshift	c.2349dupA	p.Glu784fs
2:25239188:CT:C	rs1255237375	frameshift	c.2349delA	p.Glu784fs
2:25239191:T:TG	NA	frameshift	c.2346dupC	p.Lys783fs
2:25239191:TG:T	NA	frameshift	c.2346delC	p.Glu784fs
2:25239193:G:A	NA	missense	c.2345C>T	p.Ala782Val
2:25239194:C:CA TCA	NA	frameshift	c.2340_2343dupTG AT	p.Ala782fs
2:25239194:C:T	NA	missense	c.2344G>A	p.Ala782Thr
2:25239194:CATC A:C	NA	frameshift	c.2340_2343delTG AT	p.Ile780fs
2:25239195:AT:A	NA	frameshift	c.2342delA	p.Asp781fs
2:25239197:CA:C	NA	frameshift	c.2340delT	p.Ile780fs
2:25239198:A:C	NA	missense	c.2340T>G	p.Ile780Met
2:25239198:AATC ATC:A	NA	inframe_indel	c.2334_2339delGA TGAT	p.Met779_Ile780 del
2:25239198:AATC ATCACAG:A	NA	frameshift	c.2330_2339delCT GTGATGAT	p.Pro777fs
2:25239199:A:C	rs370751539	missense	c.2339T>G	p.Ile780Ser
2:25239199:A:G	rs370751539	missense	c.2339T>C	p.Ile780Thr
2:25239199:A:T	rs370751539	missense	c.2339T>A	p.Ile780Asn
2:25239201:C:CA TCA	NA	frameshift	c.2333_2336dupTG AT	p.Met779fs
2:25239201:CA:C	NA	frameshift	c.2336delT	p.Met779fs
2:25239202:A:G	NA	missense	c.2336T>C	p.Met779Thr
2:25239202:AT:A	NA	frameshift	c.2335delA	p.Met779fs
2:25239203:TCAC AGGGTTGGACTA CAAAACAG:T	NA	splice_acceptor	c.2323- 11_2334delCTGTTT TGTAGTCCAACCCT GTG	p.Ser775_Met779 del
2:25239204:C:CA	NA	frameshift	c.2333dupT	p.Met779fs
2:25239204:CA:C	NA	frameshift	c.2333delT	p.Val778fs

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2:25239204:CACA GGGTTGGACTAC AA:C	NA	frameshift	c.2323- 6_2333delTTGTAG TCCAACCCTGT	p.Ser775fs
2:25239204:CACA GGGTTGGACTAC AAA:C	NA	frameshift	c.2323- 7_2333delTTTGTA GTCCAACCCTGT	p.Ser775fs
2:25239205:A:C	rs979932565	missense	c.2333T>G	p.Val778Gly
2:25239205:A:G	NA	missense	c.2333T>C	p.Val778Ala
2:25239205:A:T	NA	missense	c.2333T>A	p.Val778Glu
2:25239205:AC:A	NA	frameshift	c.2332delG	p.Val778fs
2:25239206:C:A	NA	missense	c.2332G>T	p.Val778Leu
2:25239206:C:T	NA	missense	c.2332G>A	p.Val778Met
2:25239207:AG:A	rs775989176	frameshift	c.2330delC	p.Pro777fs
2:25239208:G:A	rs752626029	missense	c.2330C>T	p.Pro777Leu
2:25239208:G:C	rs752626029	missense	c.2330C>G	p.Pro777Arg
2:25239208:G:T	rs752626029	missense	c.2330C>A	p.Pro777His
2:25239209:G:A	rs1172777420	missense	c.2329C>T	p.Pro777Ser
2:25239210:GT:G	NA	frameshift	c.2327delA	p.Asn776fs
2:25239212:T:TG	NA	frameshift	c.2325dupC	p.Asn776fs
2:25239212:TG:T	NA	frameshift	c.2325delC	p.Asn776fs
2:25239214:G:A	NA	missense	c.2324C>T	p.Ser775Phe
2:25239214:GA:G	NA	frameshift	c.2323delT	p.Ser775fs
2:25239215:A:G	rs74708853	missense	c.2323T>C	p.Ser775Pro
2:25239215:A:T	NA	missense	c.2323T>A	p.Ser775Thr
2:25239216:C:G	NA	splice_acceptor	c.2323-1G>C	
2:25239216:C:T	rs1324347218	splice_acceptor	c.2323-1G>A	
2:25239217:T:A	rs765045799	splice_acceptor	c.2323-2A>T	
2:25239217:T:C	rs765045799	splice_acceptor	c.2323-2A>G	
2:25239217:T:G	NA	splice_acceptor	c.2323-2A>C	

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2:25240299:TACC TCG:T	NA	frameshift	c.2319_2322+2del CGAGGT	p.Glu774fs
2:25240300:A:AC CT	NA	splice_donor	c.2321_2322+1dup AGG	
2:25240300:A:C	NA	splice_donor	c.2322+2T>G	
2:25240300:A:G	NA	splice_donor	c.2322+2T>C	
2:25240300:AC:A	NA	splice_donor	c.2322+1delG	
2:25240300:ACCT CGAGAAATCGCG AGAT:A	NA	splice_donor	c.2305_2322+1del ATCTCGCGATTCT CGAGG	p.Ile769_Glu774d el
2:25240300:ACCT CGAGAAATCGCG AGATGTC:A	NA	splice_donor	c.2302_2322+1del GACATCTCGCGATT TCTCGAGG	p.Asp768_Glu774 del
2:25240301:C:A	rs903011938	splice_donor	c.2322+1G>T	
2:25240301:C:CC TCGAGAAAT	NA	frameshift	c.2313_2322dupAT TTCTCGAG	p.Ser775fs
2:25240301:C:G	NA	splice_donor	c.2322+1G>C	
2:25240301:C:T	rs903011938	splice_donor	c.2322+1G>A	
2:25240301:CCT: C	NA	frameshift	c.2321_2322delAG	p.Glu774fs
2:25240301:CCTC GA:C	NA	frameshift	c.2318_2322delTC GAG	p.Glu774fs
2:25240302:C:A	rs753017271	missense	c.2322G>T	p.Glu774Asp
2:25240302:C:CT CGAGAAA	NA	frameshift	c.2314_2321dupTT TCTCGA	p.Glu774fs
2:25240302:C:G	rs753017271	missense	c.2322G>C	p.Glu774Asp
2:25240302:CTCG A:C	NA	frameshift	c.2318_2321delTC GA	p.Leu773fs
2:25240303:T:A	NA	missense	c.2321A>T	p.Glu774Val
2:25240303:T:C		missense	c.2321A>G	p.Glu774Gly
2:25240303:T:G	NA	missense	c.2321A>C	p.Glu774Ala
2:25240303:T:TC	NA	frameshift	c.2320dupG	p.Glu774fs
2:25240303:T:TC GAGAAATCG	rs761980712	frameshift	c.2311_2320dupCG ATTTCTCG	p.Glu774fs

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2:25240303:TC:T	NA	frameshift	c.2320delG	p.Glu774fs
2:25240304:C:A	NA	stop_gained	c.2320G>T	p.Glu774*
2:25240304:C:CG	NA	frameshift	c.2319dupC	p.Glu774fs
2:25240304:C:T	rs760624806	missense	c.2320G>A	p.Glu774Lys
2:25240304:CGA GAA:C	NA	frameshift	c.2315_2319delTTC TC	p.Phe772fs
2:25240305:G:GA GAAATCGCGAGA TGTCCCTCTTGTC ACT	NA	frameshift	c.2290_2318dupA GTGACAAGAGGGA CATCTCGCGATTTC T	p.Glu774fs
2:25240305:GAG A:G	NA	inframe_indel	c.2316_2318delTCT	p.Leu773del
2:25240306:A:C	rs764303486	missense	c.2318T>G	p.Leu773Arg
2:25240306:A:G	NA	missense	c.2318T>C	p.Leu773Pro
2:25240306:A:T	NA	missense	c.2318T>A	p.Leu773His
2:25240306:AG:A	NA	frameshift	c.2317delC	p.Leu773fs
2:25240307:G:A	NA	missense	c.2317C>T	p.Leu773Phe
2:25240307:G:C	NA	missense	c.2317C>G	p.Leu773Val
2:25240307:G:GA A	NA	frameshift	c.2315_2316dupTT	p.Leu773fs
2:25240307:G:T	rs754336556	missense	c.2317C>A	p.Leu773Ile
2:25240307:GA:G	NA	frameshift	c.2316delT	p.Leu773fs
2:25240307:GAA: G	NA	frameshift	c.2315_2316delTT	p.Phe772fs
2:25240308:A:AA ATCGCGAGATGTC CCTCTTGTCACCTA ACGCCC	NA	inframe_indel	c.2283_2315dupG GGCGTTAGTGACAA GAGGGACATCTCGC GATT	p.Arg771_Phe772 insLeuGlyValSerA spLysArgAspIleSer Arg
2:25240308:A:C	NA	missense	c.2316T>G	p.Phe772Leu
2:25240308:A:T	NA	missense	c.2316T>A	p.Phe772Leu
2:25240308:AAAT C:A	NA	frameshift	c.2312_2315delGA TT	p.Arg771fs
2:25240309:A:AA TCGCGAGATGTCC	NA	frameshift	c.2272_2314dupGT GGTGGCCATGGGC	p.Phe772fs

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CTCTTGCTACTAA CGCCCATGGCCAC CAC			GTTAGTGACAAGA GGGACATCTCGCGA T	
2:25240309:A:AT C	NA	frameshift	c.2314_2315insGA	p.Phe772fs
2:25240309:A:C	NA	missense	c.2315T>G	p.Phe772Cys
2:25240309:A:G	NA	missense	c.2315T>C	p.Phe772Ser
2:25240309:AATC GCGAG:A	NA	frameshift	c.2307_2314delCT CGCGAT	p.Phe772fs
2:25240310:A:AT CGCGAGATGTCCC TCTTG	NA	frameshift	c.2295_2313dupCA AGAGGGACATCTC GCGA	p.Phe772fs
2:25240310:A:G	NA	missense	c.2314T>C	p.Phe772Leu
2:25240310:A:T	NA	missense	c.2314T>A	p.Phe772Ile
2:25240310:AT:A	NA	frameshift	c.2313delA	p.Leu773fs
2:25240311:T:TC GCGAGATGTCCCT CTTGCTACTAA	NA	frameshift	c.2288_2312dupTT AGTGACAAGAGGG ACATCTCGCG	p.Phe772fs
2:25240311:TC:T	NA	frameshift	c.2312delG	p.Arg771fs
2:25240311:TCGC G:T	NA	frameshift	c.2309_2312delCG CG	p.Ser770fs
2:25240312:C:A	rs757823678	missense	c.2312G>T	p.Arg771Leu
2:25240312:C:CG ATTT	NA	frameshift	c.2311_2312insAA ATC	p.Arg771fs
2:25240312:C:G	NA	missense	c.2312G>C	p.Arg771Pro
2:25240312:C:T	rs757823678	missense	c.2312G>A	p.Arg771Gln
2:25240312:CG:C	NA	frameshift	c.2311delC	p.Arg771fs
2:25240312:CGC GAGATGTCCCTCT TGTCCTA:C	NA	frameshift	c.2289_2311delTA GTGACAAGAGGGA CATCTCGC	p.Ser764fs
2:25240313:G:A	rs779626155	stop_gained	c.2311C>T	p.Arg771*
2:25240313:G:C	NA	missense	c.2311C>G	p.Arg771Gly
2:25240313:G:GC	NA	frameshift	c.2310dupG	p.Arg771fs

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2:25240313:GCG AGATGTCCCTCTT GTCA:G	NA	frameshift	c.2292_2310delTG ACAAGAGGGACAT CTCG	p.Lys766fs
2:25240314:C:CG AG	NA	inframe_indel	c.2307_2309dupCT C	p.Ser770dup
2:25240314:CGA: C	NA	frameshift	c.2308_2309delTC	p.Ser770fs
2:25240315:G:A	rs758845779	missense	c.2309C>T	p.Ser770Leu
2:25240315:G:C	rs758845779	missense	c.2309C>G	p.Ser770Trp
2:25240315:G:GA	NA	frameshift	c.2308dupT	p.Ser770fs
2:25240315:G:T	rs758845779	stop_gained	c.2309C>A	p.Ser770*
2:25240315:GA:G	NA	frameshift	c.2308delT	p.Ser770fs
2:25240316:A:C	NA	missense	c.2308T>G	p.Ser770Ala
2:25240316:A:G	rs1275276226	missense	c.2308T>C	p.Ser770Pro
2:25240316:AG:A	NA	frameshift	c.2307delC	p.Ser770fs
2:25240317:G:C	NA	missense	c.2307C>G	p.Ile769Met
2:25240317:G:GT	NA	frameshift	c.2306_2307insA	p.Ser770fs
2:25240317:GA:G	NA	frameshift	c.2306delT	p.Ile769fs
2:25240318:A:C	rs746704362	missense	c.2306T>G	p.Ile769Ser
2:25240318:A:G	rs746704362	missense	c.2306T>C	p.Ile769Thr
2:25240318:A:T	rs746704362	missense	c.2306T>A	p.Ile769Asn
2:25240318:AT:A	NA	frameshift	c.2305delA	p.Ile769fs
2:25240319:T:C	NA	missense	c.2305A>G	p.Ile769Val
2:25240319:T:G	NA	missense	c.2305A>C	p.Ile769Leu
2:25240320:G:C	NA	missense	c.2304C>G	p.Asp768Glu
2:25240320:G:T	NA	missense	c.2304C>A	p.Asp768Glu
2:25240320:GT:G	NA	frameshift	c.2303delA	p.Asp768fs
2:25240321:T:A	NA	missense	c.2303A>T	p.Asp768Val
2:25240321:T:C	rs1181120114	missense	c.2303A>G	p.Asp768Gly
2:25240321:TC:T	NA	frameshift	c.2302delG	p.Asp768fs
2:25240322:C:A	rs767983115	missense	c.2302G>T	p.Asp768Tyr
2:25240322:C:G	NA	missense	c.2302G>C	p.Asp768His
2:25240322:C:T	NA	missense	c.2302G>A	p.Asp768Asn

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2:25240323:C:CC T	NA	frameshift	c.2299_2300dupA G	p.Asp768fs
2:25240324:CT:C	NA	frameshift	c.2299delA	p.Arg767fs
2:25240324:CTCT TG:C	NA	frameshift	c.2295_2299delCA AGA	p.Asp765fs
2:25240325:T:C	NA	missense	c.2299A>G	p.Arg767Gly
2:25240325:TC:T	NA	frameshift	c.2298delG	p.Arg767fs
2:25240326:C:A	NA	missense	c.2298G>T	p.Lys766Asn
2:25240326:C:G	NA	missense	c.2298G>C	p.Lys766Asn
2:25240326:CT:C	NA	frameshift	c.2297delA	p.Lys766fs
2:25240326:CTT:C	rs750475955	frameshift	c.2296_2297delAA	p.Lys766fs
2:25240327:TTG: T	NA	frameshift	c.2295_2296delCA	p.Asp765fs
2:25240328:T:A	NA	stop_gained	c.2296A>T	p.Lys766*
2:25240328:T:C	NA	missense	c.2296A>G	p.Lys766Glu
2:25240328:T:TA	NA	frameshift	c.2295_2296insT	p.Lys766fs
2:25240328:TG:T	NA	frameshift	c.2295delC	p.Asp765fs
2:25240329:G:GT	NA	frameshift	c.2294dupA	p.Asp765fs
2:25240330:T:C	NA	missense	c.2294A>G	p.Asp765Gly
2:25240331:C:A	NA	missense	c.2293G>T	p.Asp765Tyr
2:25240332:AC:A	NA	frameshift	c.2291delG	p.Ser764fs
2:25240333:C:CT	NA	frameshift	c.2290dupA	p.Ser764fs
2:25240333:CT:C	NA	frameshift	c.2290delA	p.Ser764fs
2:25240334:T:TA	NA	frameshift	c.2289dupT	p.Ser764fs
2:25240334:TA:T	NA	frameshift	c.2289delT	p.Ser764fs
2:25240336:A:T	rs1178258952	missense	c.2288T>A	p.Val763Asp
2:25240336:AC:A	NA	frameshift	c.2287delG	p.Val763fs
2:25240337:C:CA	NA	frameshift	c.2286_2287insT	p.Val763fs
2:25240337:C:CG TTA	NA	frameshift	c.2286_2287insTA AC	p.Val763fs
2:25240337:C:T	rs1369746569	missense	c.2287G>A	p.Val763Ile
2:25240338:GC:G	NA	frameshift	c.2285delG	p.Gly762fs

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2:25240338:GCCC ATGGCCA:G	NA	frameshift	c.2276_2285delTG GCCATGGG	p.Val759fs
2:25240342:A:C	NA	missense	c.2282T>G	p.Met761Arg
2:25240342:A:G	rs963262801	missense	c.2282T>C	p.Met761Thr
2:25240342:ATG GCCACCACAT:A	NA	inframe_indel	c.2270_2281delAT GTGGTGGCCA	p.Asn757_Ala760 del
2:25240343:T:C	rs780695582	missense	c.2281A>G	p.Met761Val
2:25240343:T:G	NA	missense	c.2281A>C	p.Met761Leu
2:25240343:T:TG	NA	frameshift	c.2280dupC	p.Met761fs
2:25240343:TG:T	NA	frameshift	c.2280delC	p.Met761fs
2:25240345:G:A	NA	missense	c.2279C>T	p.Ala760Val
2:25240345:G:C	NA	missense	c.2279C>G	p.Ala760Gly
2:25240345:G:GC	NA	frameshift	c.2278dupG	p.Ala760fs
2:25240345:GC:G	NA	frameshift	c.2278delG	p.Ala760fs
2:25240345:GCCA :G	NA	inframe_indel	c.2276_2278delTG G	p.Val759del
2:25240346:CCAC CACATTCT:C	NA	frameshift	c.2267_2277delAG AATGTGGTG	p.Glu756fs
2:25240347:CA:C	NA	frameshift	c.2276delT	p.Val759fs
2:25240348:AC:A	NA	frameshift	c.2275delG	p.Val759fs
2:25240349:C:G	NA	missense	c.2275G>C	p.Val759Leu
2:25240349:C:T	NA	missense	c.2275G>A	p.Val759Met
2:25240349:CCA: C	NA	frameshift	c.2273_2274delTG	p.Val758fs
2:25240352:C:G	NA	missense	c.2272G>C	p.Val758Leu
2:25240352:C:T	NA	missense	c.2272G>A	p.Val758Met
2:25240352:CA:C	NA	frameshift	c.2271delT	p.Asn757fs
2:25240352:CATT CTCAAAGAGCCAG :C	NA	frameshift	c.2256_2271delCT GGCTCTTTGAGAAT	p.Phe752fs
2:25240353:A:C	NA	missense	c.2271T>G	p.Asn757Lys
2:25240353:A:T	NA	missense	c.2271T>A	p.Asn757Lys
2:25240353:AT:A	NA	frameshift	c.2270delA	p.Asn757fs

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2:25240353:ATTC TCAAAGAGCCAG AAGAAGGG:A	NA	frameshift	c.2248_2270delCC CTTCTTCTGGCTCTT TGAGAA	p.Pro750fs
2:25240354:T:A	rs747716263	missense	c.2270A>T	p.Asn757Ile
2:25240354:T:C	NA	missense	c.2270A>G	p.Asn757Ser
2:25240354:T:TTC	NA	frameshift	c.2268_2269dupG A	p.Asn757fs
2:25240355:T:A	NA	missense	c.2269A>T	p.Asn757Tyr
2:25240356:C:A	rs769718039	missense	c.2268G>T	p.Glu756Asp
2:25240356:C:G	NA	missense	c.2268G>C	p.Glu756Asp
2:25240356:CTCA AAGAGCCAGAAG AA:C	NA	frameshift	c.2251_2267delTTC TTCTGGCTCTTTGA	p.Phe751fs
2:25240357:T:C	NA	missense	c.2267A>G	p.Glu756Gly
2:25240357:T:TC A	NA	frameshift	c.2265_2266dupTG	p.Glu756fs
2:25240358:C:A	NA	stop_gained	c.2266G>T	p.Glu756*
2:25240358:C:CA	rs1386755076	frameshift	c.2265dupT	p.Glu756fs
2:25240358:C:T	rs1418213272	missense	c.2266G>A	p.Glu756Lys
2:25240358:CA:C	rs752551332	frameshift	c.2265delT	p.Phe755fs
2:25240358:CAA: C	rs755800729	frameshift	c.2264_2265delTT	p.Phe755fs
2:25240358:CAAA GA:C	NA	frameshift	c.2261_2265delTCT TT	p.Leu754fs
2:25240359:A:AT	NA	frameshift	c.2264_2265insA	p.Phe755fs
2:25240359:A:C	NA	missense	c.2265T>G	p.Phe755Leu
2:25240359:A:T	NA	missense	c.2265T>A	p.Phe755Leu
2:25240360:A:C	NA	missense	c.2264T>G	p.Phe755Cys
2:25240360:A:G	rs536841393	missense	c.2264T>C	p.Phe755Ser
2:25240360:AAG: A	NA	frameshift	c.2262_2263delICT	p.Phe755fs
2:25240360:AAG AGCC:A	NA	inframe_indel	c.2258_2263delIGG CTCT	p.Trp753_Leu754 del

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2:25240360:AAG AGCCAGAAGAAG: A	NA	frameshift	c.2250_2263delCTT CTTCTGGCTCT	p.Phe751fs
2:25240361:A:C	NA	missense	c.2263T>G	p.Phe755Val
2:25240361:A:G	NA	missense	c.2263T>C	p.Phe755Leu
2:25240361:A:T	rs369230209	missense	c.2263T>A	p.Phe755Ile
2:25240361:AGA GCCAGAAGAAGG GGC:A	NA	frameshift	c.2246_2262delGC CCCTTCTTCTGGCTC	p.Arg749fs
2:25240362:GA:G	NA	frameshift	c.2261delT	p.Leu754fs
2:25240362:GAG CC:G	NA	frameshift	c.2258_2261delGG CT	p.Trp753fs
2:25240363:A:C	rs762636955	missense	c.2261T>G	p.Leu754Arg
2:25240363:A:G	NA	missense	c.2261T>C	p.Leu754Pro
2:25240363:A:T	NA	missense	c.2261T>A	p.Leu754His
2:25240364:G:C	NA	missense	c.2260C>G	p.Leu754Val
2:25240364:GCCA GAAGAAGGGGCG AT:G	NA	frameshift	c.2243_2259delAT CGCCCCCTTCTCTG G	p.Asp748fs
2:25240365:C:A	NA	missense	c.2259G>T	p.Trp753Cys
2:25240365:C:CTT	NA	frameshift	c.2258_2259insAA	p.Trp753fs
2:25240365:C:G	rs770785915	missense	c.2259G>C	p.Trp753Cys
2:25240365:C:T	NA	stop_gained	c.2259G>A	p.Trp753*
2:25240365:CCAG :C	NA	inframe_indel	c.2256_2258delCT G	p.Phe752_Trp753 delinsLeu
2:25240366:C:A	rs775537912	missense	c.2258G>T	p.Trp753Leu
2:25240366:C:CA G	NA	frameshift	c.2256_2257dupCT	p.Trp753fs
2:25240366:C:G		missense	c.2258G>C	p.Trp753Ser
2:25240366:C:T		stop_gained	c.2258G>A	p.Trp753*
2:25240366:CAG: C	NA	frameshift	c.2256_2257delCT	p.Phe752fs
2:25240366:CAG A:C	rs749132507	inframe_indel	c.2255_2257delTCT	p.Phe752del

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2:25240366:CAG AAGAAGGGGCG: C	NA	frameshift	c.2245_2257delCG CCCCTTCTTCT	p.Arg749fs
2:25240367:A:C	NA	missense	c.2257T>G	p.Trp753Gly
2:25240367:A:G	NA	missense	c.2257T>C	p.Trp753Arg
2:25240367:A:T	rs1189107128	missense	c.2257T>A	p.Trp753Arg
2:25240367:AG:A	rs770968422	frameshift	c.2256delC	p.Trp753fs
2:25240367:AGA AG:A	NA	frameshift	c.2253_2256delCTT C	p.Phe752fs
2:25240367:AGA AGAAGGGGC:A	NA	frameshift	c.2246_2256delGC CCCTTCTTC	p.Arg749fs
2:25240368:G:C	rs760959622	missense	c.2256C>G	p.Phe752Leu
2:25240368:G:GC	NA	frameshift	c.2255_2256insG	p.Phe752fs
2:25240368:G:T	NA	missense	c.2256C>A	p.Phe752Leu
2:25240368:GA:G	NA	frameshift	c.2255delT	p.Phe752fs
2:25240368:GAA: G	NA	frameshift	c.2254_2255delTT	p.Phe752fs
2:25240369:A:AA GAAGGGGCGATC	NA	frameshift	c.2242_2254dupG ATCGCCCCTTCT	p.Phe752fs
2:25240369:A:C	rs764464925	missense	c.2255T>G	p.Phe752Cys
2:25240369:A:G	NA	missense	c.2255T>C	p.Phe752Ser
2:25240369:AAG: A	NA	frameshift	c.2253_2254delCT	p.Phe752fs
2:25240369:AAG AAGGGG:A	NA	frameshift	c.2247_2254delCC CCTTCT	p.Pro750fs
2:25240370:A:AG AAGGGGCTTT	NA	frameshift	c.2253_2254insAA AGCCCCTTC	p.Phe752fs
2:25240370:A:C	NA	missense	c.2254T>G	p.Phe752Val
2:25240370:A:G	rs776844126	missense	c.2254T>C	p.Phe752Leu
2:25240370:A:T		missense	c.2254T>A	p.Phe752Ile
2:25240370:AG:A	NA	frameshift	c.2253delC	p.Phe752fs
2:25240370:AGA AGGGG:A	NA	frameshift	c.2247_2253delCC CCTTC	p.Pro750fs
2:25240371:G:C	rs762020470	missense	c.2253C>G	p.Phe751Leu

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2:25240371:G:GA	NA	frameshift	c.2252dupT	p.Phe752fs
2:25240371:G:GT	NA	frameshift	c.2252_2253insA	p.Phe751fs
2:25240371:G:T	NA	missense	c.2253C>A	p.Phe751Leu
2:25240371:GA:G	NA	frameshift	c.2252delT	p.Phe751fs
2:25240372:A:C	rs765813304	missense	c.2252T>G	p.Phe751Cys
2:25240372:A:G	rs765813304	missense	c.2252T>C	p.Phe751Ser
2:25240373:A:AG	rs1195416744	frameshift	c.2250dupC	p.Phe751fs
2:25240373:A:C	NA	missense	c.2251T>G	p.Phe751Val
2:25240373:A:G	NA	missense	c.2251T>C	p.Phe751Leu
2:25240373:A:T	rs750962348	missense	c.2251T>A	p.Phe751Ile
2:25240373:AG:A	NA	frameshift	c.2250delC	p.Phe751fs
2:25240374:GGG GC:G	NA	frameshift	c.2246_2249delG CC	p.Arg749fs
2:25240375:G:C	NA	missense	c.2249C>G	p.Pro750Arg
2:25240375:G:GG GCGATCATCTCCC TCCTT	NA	frameshift	c.2230_2248dupAA GGAGGGAGATGAT CGCC	p.Pro750fs
2:25240375:GGG CGATCAT:G	NA	inframe_indel	c.2240_2248delAT GATCGCC	p.Asp747_Pro750 delinsAla
2:25240376:G:A	NA	missense	c.2248C>T	p.Pro750Ser
2:25240376:G:C	NA	missense	c.2248C>G	p.Pro750Ala
2:25240376:G:GG CGATCATCTCCC	NA	frameshift	c.2235_2247dupG GGAGATGATCGC	p.Pro750fs
2:25240376:GGC: G	NA	frameshift	c.2246_2247delGC	p.Arg749fs
2:25240377:G:GA	NA	frameshift	c.2246_2247insT	p.Phe751fs
2:25240377:G:GC	NA	frameshift	c.2246dupG	p.Phe751fs
2:25240377:G:GC GATCATCTCCCTC CTTGGGCCTCTGG CT	NA	frameshift	c.2246_2247insAG CCAGAGGCCCAAG GAGGGAGATGATC G	p.Pro750fs
2:25240377:GC:G	NA	frameshift	c.2246delG	p.Arg749fs
2:25240377:GCG AT:G	NA	frameshift	c.2243_2246delAT CG	p.Asp748fs

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2:25240377:GCG ATCATCTCCCTCCT TGGGC:G	NA	inframe_indel	c.2226_2246delGC CCAAGGAGGGAGA TGATCG	p.Pro743_Arg749 del
2:25240378:C:A	rs34843713	missense	c.2246G>T	p.Arg749Leu
2:25240378:C:G	rs34843713	missense	c.2246G>C	p.Arg749Pro
2:25240378:C:T	rs34843713	missense	c.2246G>A	p.Arg749His
2:25240378:CG:C	NA	frameshift	c.2245delC	p.Arg749fs
2:25240378:CGAT CA:C	NA	frameshift	c.2241_2245delTG ATC	p.Asp747fs
2:25240379:G:A	rs754613602	missense	c.2245C>T	p.Arg749Cys
2:25240379:G:C	rs754613602	missense	c.2245C>G	p.Arg749Gly
2:25240379:G:GT	NA	frameshift	c.2244_2245insA	p.Arg749fs
2:25240379:G:T	NA	missense	c.2245C>A	p.Arg749Ser
2:25240379:GA:G	NA	frameshift	c.2244delT	p.Arg749fs
2:25240379:GATC A:G	NA	frameshift	c.2241_2244delTG AT	p.Asp748fs
2:25240380:AT:A	NA	frameshift	c.2243delA	p.Asp748fs
2:25240381:T:TC	NA	frameshift	c.2242dupG	p.Asp748fs
2:25240381:T:TC ATCTCCCTCCTTG GGCCG	NA	frameshift	c.2224_2242dupCG GCCCAAGGAGGGA GATG	p.Asp748fs
2:25240381:TC:T	NA	frameshift	c.2242delG	p.Asp748fs
2:25240383:A:AT C	NA	frameshift	c.2239_2240dupG A	p.Asp747fs
2:25240384:T:C	NA	missense	c.2240A>G	p.Asp747Gly
2:25240384:T:G	NA	missense	c.2240A>C	p.Asp747Ala
2:25240384:TCTC CCTC:T	NA	frameshift	c.2233_2239delGA GGGAG	p.Glu745fs
2:25240385:C:A	NA	missense	c.2239G>T	p.Asp747Tyr
2:25240385:CT:C	NA	frameshift	c.2238delA	p.Asp747fs
2:25240385:CTCC CTCCTTGGGCCGC GCAT:C	NA	frameshift	c.2219_2238delAT GCGCGGCCCAAGG AGGGA	p.Asp740fs
2:25240386:TC:T	NA	frameshift	c.2237delG	p.Gly746fs

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2:25240387:C:T	NA	missense	c.2237G>A	p.Gly746Glu
2:25240388:C:A	NA	stop_gained	c.2236G>T	p.Gly746*
2:25240388:C:T	rs747626849	missense	c.2236G>A	p.Gly746Arg
2:25240389:CTCC T:C	NA	frameshift	c.2231_2234delAG GA	p.Lys744fs
2:25240390:T:TCC	NA	frameshift	c.2232_2233dupG G	p.Glu745fs
2:25240390:T:TCC TTGGG	NA	frameshift	c.2227_2233dupCC CAAGG	p.Glu745fs
2:25240391:C:A	rs1328524162	stop_gained	c.2233G>T	p.Glu745*
2:25240392:C:CT	NA	frameshift	c.2231dupA	p.Glu745fs
2:25240392:CT:C	NA	frameshift	c.2231delA	p.Lys744fs
2:25240394:T:A	rs1389075270	stop_gained	c.2230A>T	p.Lys744*
2:25240394:T:C	NA	missense	c.2230A>G	p.Lys744Glu
2:25240394:T:TG	NA	frameshift	c.2229dupC	p.Lys744fs
2:25240394:T:TG GGCCGCGCATCAT GCAGGAGGCGGT AGAACTCAAAGA AGAGCCGGCCAG TGC	NA	frameshift	c.2177_2229dupGC ACTGGCCGGCTCTT CTTTGAGTTCTACC GCCTCCTGCATGAT GCGCGGCC	p.Lys744fs
2:25240394:T:TTA	NA	frameshift	c.2229_2230insTA	p.Lys744fs
2:25240394:TG:T	NA	frameshift	c.2229delC	p.Lys744fs
2:25240394:TGG GCC:T	NA	frameshift	c.2225_2229delGG CCC	p.Arg742fs
2:25240396:G:A	rs755673156	missense	c.2228C>T	p.Pro743Leu
2:25240396:G:C	rs755673156	missense	c.2228C>G	p.Pro743Arg
2:25240396:G:GG CCGC	NA	frameshift	c.2223_2227dupGC GGC	p.Pro743fs
2:25240396:G:T	NA	missense	c.2228C>A	p.Pro743His
2:25240397:G:A	rs1433353413	missense	c.2227C>T	p.Pro743Ser
2:25240397:G:C	rs1433353413	missense	c.2227C>G	p.Pro743Ala
2:25240397:G:GC	NA	frameshift	c.2226dupG	p.Pro743fs
2:25240397:GC:G	rs778606920	frameshift	c.2226delG	p.Lys744fs

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2:25240398:C:CC G	rs745364706	frameshift	c.2224_2225dupCG	p.Pro743fs
2:25240398:CCG: C	NA	frameshift	c.2224_2225delCG	p.Arg742fs
2:25240398:CCGC G:C	NA	frameshift	c.2222_2225delCG CG	p.Ala741fs
2:25240398:CCGC GCATCATG:C	NA	frameshift	c.2215_2225delCA TGATGCGCG	p.His739fs
2:25240399:C:CG	NA	frameshift	c.2224dupC	p.Arg742fs
2:25240399:C:CT	NA	frameshift	c.2224_2225insA	p.Arg742fs
2:25240399:C:G	rs749282210	missense	c.2225G>C	p.Arg742Pro
2:25240399:C:T	NA	missense	c.2225G>A	p.Arg742Gln
2:25240399:CG:C	NA	frameshift	c.2224delC	p.Arg742fs
2:25240399:CGC GCA:C	NA	frameshift	c.2220_2224delTG CGC	p.Asp740fs
2:25240399:CGC GCATCATG:C	NA	frameshift	c.2215_2224delCA TGATGCGCG	p.His739fs
2:25240399:CGC GCATCATGCA:C	NA	inframe_indel	c.2213_2224delTG CATGATGCGCG	p.Leu738_Ala741 del
2:25240399:CGC GCATCATGCAGGA G:C	NA	frameshift	c.2209_2224delCT CCTGCATGATGCGCG	p.Leu737fs
2:25240400:G:C	NA	missense	c.2224C>G	p.Arg742Gly
2:25240402:G:A	rs770568549	missense	c.2222C>T	p.Ala741Val
2:25240402:G:C	NA	missense	c.2222C>G	p.Ala741Gly
2:25240402:G:GC	NA	frameshift	c.2221dupG	p.Ala741fs
2:25240402:G:T	rs770568549	missense	c.2222C>A	p.Ala741Glu
2:25240403:C:CA	NA	frameshift	c.2220dupT	p.Ala741fs
2:25240403:C:CA TCATGCAGG	NA	frameshift	c.2211_2220dupCC TGCATGAT	p.Ala741fs
2:25240403:C:G	NA	missense	c.2221G>C	p.Ala741Pro
2:25240403:C:T	NA	missense	c.2221G>A	p.Ala741Thr
2:25240403:CATC A:C	NA	frameshift	c.2217_2220delTG AT	p.His739fs

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2:25240404:A:AA CG	NA	inframe_indel	c.2219_2220insCG T	p.Asp740_Ala741i nsVal
2:25240406:C:CA	NA	frameshift	c.2217dupT	p.Asp740fs
2:25240407:A:AT GCAGGAGGCGGT AGAACTCAAAGA AGAGCCGGCCAG TGCCCTC	NA	frameshift	c.2174- 1_2216dupGAGGG CACTGGCCGGCTCT TCTTTGAGTTCTACC GCCTCCTGCA	p.His739fs
2:25240408:T:C	NA	missense	c.2216A>G	p.His739Arg
2:25240408:T:G	rs1053660507	missense	c.2216A>C	p.His739Pro
2:25240409:G:A	NA	missense	c.2215C>T	p.His739Tyr
2:25240410:CAG G:C	NA	inframe_indel	c.2211_2213delCC T	p.Leu738del
2:25240411:A:AG	NA	frameshift	c.2212dupC	p.Leu738fs
2:25240411:A:C	rs745714537	missense	c.2213T>G	p.Leu738Arg
2:25240411:A:G	NA	missense	c.2213T>C	p.Leu738Pro
2:25240411:A:T	rs745714537	missense	c.2213T>A	p.Leu738Gln
2:25240411:AG:A	NA	frameshift	c.2212delC	p.Leu738fs
2:25240412:G:C	rs1374722278	missense	c.2212C>G	p.Leu738Val
2:25240412:GGA GGCGGTAGAACT: G	NA	frameshift	c.2198_2211delAG TTCTACCGCCTC	p.Glu733fs
2:25240413:GA:G	NA	frameshift	c.2210delT	p.Leu737fs
2:25240414:A:C	rs768964533	missense	c.2210T>G	p.Leu737Arg
2:25240414:A:G	NA	missense	c.2210T>C	p.Leu737Pro
2:25240414:A:T	NA	missense	c.2210T>A	p.Leu737His
2:25240414:AG:A	NA	frameshift	c.2209delC	p.Leu737fs
2:25240415:G:A	rs1340822377	missense	c.2209C>T	p.Leu737Phe
2:25240416:GCG GTAGAACT:G	NA	frameshift	c.2198_2207delAG TTCTACCG	p.Glu733fs
2:25240416:GCG GTAGAACTCAAAG A:G	NA	frameshift	c.2192_2207delTCT TTGAGTTCTACCG	p.Phe731fs
2:25240417:C:A	NA	missense	c.2207G>T	p.Arg736Leu

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2:25240417:C:G	NA	missense	c.2207G>C	p.Arg736Pro
2:25240417:C:T	rs139293773	missense	c.2207G>A	p.Arg736His
2:25240418:G:A	rs761934754	missense	c.2206C>T	p.Arg736Cys
2:25240418:G:C	rs761934754	missense	c.2206C>G	p.Arg736Gly
2:25240418:G:T	rs761934754	missense	c.2206C>A	p.Arg736Ser
2:25240419:G:C	rs765439043	stop_gained	c.2205C>G	p.Tyr735*
2:25240419:G:GT	NA	frameshift	c.2204dupA	p.Tyr735fs
2:25240419:G:T	NA	stop_gained	c.2205C>A	p.Tyr735*
2:25240419:GT:G	NA	frameshift	c.2204delA	p.Tyr735fs
2:25240420:T:A	NA	missense	c.2204A>T	p.Tyr735Phe
2:25240420:T:C	rs147828672	missense	c.2204A>G	p.Tyr735Cys
2:25240420:T:G	rs147828672	missense	c.2204A>C	p.Tyr735Ser
2:25240420:T:TA GAACTCAAAG	NA	frameshift	c.2193_2203dupCT TTGAGTTCT	p.Tyr735fs
2:25240420:TAG: T		frameshift	c.2202_2203delCT	p.Phe734fs
2:25240420:TAGA ACTC:T	NA	frameshift	c.2197_2203delGA GTTCT	p.Glu733fs
2:25240421:A:AG AACTCAAAGAAG AGCCGGCCAGTG CCCTCT	NA	frameshift	c.2174- 2_2202dupAGAGG GCACTGGCCGGCTC TTCTTTGAGTTC	p.Tyr735fs
2:25240421:A:C	NA	missense	c.2203T>G	p.Tyr735Asp
2:25240421:A:G	NA	missense	c.2203T>C	p.Tyr735His
2:25240421:A:T	NA	missense	c.2203T>A	p.Tyr735Asn
2:25240421:AG:A	NA	frameshift	c.2202delC	p.Tyr735fs
2:25240421:AGA ACTCAAAGAAG:A	NA	frameshift	c.2190_2202delCTT CTTTGAGTTC	p.Phe731fs
2:25240422:G:C		missense	c.2202C>G	p.Phe734Leu
2:25240422:G:GA	NA	frameshift	c.2201dupT	p.Tyr735fs
2:25240422:G:GA ACTCAAAGA	NA	frameshift	c.2192_2201dupTC TTTGAGTT	p.Tyr735fs
2:25240422:G:GA CT	NA	inframe_indel	c.2201_2202insAG T	p.Phe734delinsLeuVal

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2:25240422:G:GA T	NA	frameshift	c.2201_2202insAT	p.Phe734fs
2:25240422:G:T	rs766854284	missense	c.2202C>A	p.Phe734Leu
2:25240422:GA:G		frameshift	c.2201delT	p.Phe734fs
2:25240422:GAA CTCAAAGAAGA:G	NA	frameshift	c.2189_2201delTCT TCTTTGAGTT	p.Leu730fs
2:25240423:A:C	rs1215180755	missense	c.2201T>G	p.Phe734Cys
2:25240423:A:G	rs1215180755	missense	c.2201T>C	p.Phe734Ser
2:25240423:AACT :A	NA	inframe_indel	c.2198_2200delAG T	p.Glu733_Phe734 delinsVal
2:25240423:AACT CAAAGAAGAGCC G:A	NA	frameshift	c.2185_2200delCG GCTCTTCTTTGAGT	p.Arg729fs
2:25240424:A:C	rs751950768	missense	c.2200T>G	p.Phe734Val
2:25240424:A:G	NA	missense	c.2200T>C	p.Phe734Leu
2:25240424:A:T	NA	missense	c.2200T>A	p.Phe734Ile
2:25240425:C:CT	NA	frameshift	c.2198dupA	p.Phe734fs
2:25240425:CTCA AAGAAGAG:C	NA	frameshift	c.2188_2198delICT CTTCTTTGA	p.Leu730fs
2:25240426:T:A	NA	missense	c.2198A>T	p.Glu733Val
2:25240426:T:C	rs755376933	missense	c.2198A>G	p.Glu733Gly
2:25240426:T:G	NA	missense	c.2198A>C	p.Glu733Ala
2:25240426:T:TC	NA	frameshift	c.2197dupG	p.Glu733fs
2:25240426:TC:T	NA	frameshift	c.2197delG	p.Glu733fs
2:25240426:TCAA AGAAGAGCCGCGC: T	NA	inframe_indel	c.2183_2197delIGC CGGCTCTTCTTTG	p.Gly728_Phe732 del
2:25240427:C:A	NA	stop_gained	c.2197G>T	p.Glu733*
2:25240427:C:CA		frameshift	c.2196dupT	p.Glu733fs
2:25240427:C:CA A	rs771906613	frameshift	c.2195_2196dupTT	p.Glu733fs
2:25240427:C:CA G	NA	frameshift	c.2196_2197insCT	p.Glu733fs
2:25240427:C:G	NA	missense	c.2197G>C	p.Glu733Gln

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2:25240427:C:T	NA	missense	c.2197G>A	p.Glu733Lys
2:25240427:CA:C	NA	frameshift	c.2196delT	p.Phe732fs
2:25240427:CAA: C	rs774944755	frameshift	c.2195_2196delTT	p.Phe732fs
2:25240427:CAAA G:C	NA	frameshift	c.2193_2196delCTT T	p.Phe731fs
2:25240427:CAAA GA:C	NA	frameshift	c.2192_2196delTCT TT	p.Phe731fs
2:25240427:CAAA GAAG:C	NA	frameshift	c.2190_2196delCTT CTTT	p.Phe731fs
2:25240428:A:C	NA	missense	c.2196T>G	p.Phe732Leu
2:25240428:A:T	NA	missense	c.2196T>A	p.Phe732Leu
2:25240428:AAA G:A	rs761103716	inframe_indel	c.2193_2195delCTT	p.Phe732del
2:25240428:AAA GAAG:A	NA	inframe_indel	c.2190_2195delCTT CTT	p.Phe731_Phe73 2del
2:25240429:A:C	rs767134940	missense	c.2195T>G	p.Phe732Cys
2:25240429:A:G	NA	missense	c.2195T>C	p.Phe732Ser
2:25240429:A:T	NA	missense	c.2195T>A	p.Phe732Tyr
2:25240429:AAG: A	NA	frameshift	c.2193_2194delCT	p.Phe732fs
2:25240429:AAG AAG:A	NA	frameshift	c.2190_2194delCTT CT	p.Phe731fs
2:25240430:A:C	NA	missense	c.2194T>G	p.Phe732Val
2:25240430:A:G	NA	missense	c.2194T>C	p.Phe732Leu
2:25240430:A:T	rs149043640	missense	c.2194T>A	p.Phe732Ile
2:25240430:AG:A	NA	frameshift	c.2193delC	p.Phe732fs
2:25240430:AGA AGAGCC:A	NA	frameshift	c.2186_2193delGG CTCTTC	p.Arg729fs
2:25240431:G:C	NA	missense	c.2193C>G	p.Phe731Leu
2:25240431:G:GC	NA	frameshift	c.2192_2193insG	p.Phe731fs
2:25240431:G:GT	NA	frameshift	c.2192_2193insA	p.Phe731fs
2:25240431:G:GT T	rs768858047	frameshift	c.2192_2193insAA	p.Phe731fs

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2:25240431:G:T	NA	missense	c.2193C>A	p.Phe731Leu
2:25240431:GA:G	NA	frameshift	c.2192delT	p.Phe731fs
2:25240431:GAA: G	NA	frameshift	c.2191_2192delTT	p.Phe731fs
2:25240432:A:C	rs755585212	missense	c.2192T>G	p.Phe731Cys
2:25240432:A:G	NA	missense	c.2192T>C	p.Phe731Ser
2:25240432:A:T	NA	missense	c.2192T>A	p.Phe731Tyr
2:25240432:AAG: A	NA	frameshift	c.2190_2191delCT	p.Phe731fs
2:25240433:A:AT	NA	frameshift	c.2190_2191insA	p.Phe731fs
2:25240433:A:C	NA	missense	c.2191T>G	p.Phe731Val
2:25240433:A:G	rs143019657	missense	c.2191T>C	p.Phe731Leu
2:25240433:A:T	NA	missense	c.2191T>A	p.Phe731Ile
2:25240433:AG:A	NA	frameshift	c.2190delC	p.Phe731fs
2:25240433:AGA GCC:A	NA	frameshift	c.2186_2190delGG CTC	p.Arg729fs
2:25240434:G:GA GCC	NA	frameshift	c.2186_2189dupG GCT	p.Phe731fs
2:25240434:GA:G	NA	frameshift	c.2189delT	p.Leu730fs
2:25240434:GAG CCGGCC:G	NA	frameshift	c.2182_2189delGG CCGGCT	p.Gly728fs
2:25240435:A:AG C	NA	frameshift	c.2187_2188dupGC	p.Leu730fs
2:25240435:A:G	rs749126333	missense	c.2189T>C	p.Leu730Pro
2:25240435:AG:A	NA	frameshift	c.2188delC	p.Leu730fs
2:25240435:AGCC GGCCAGT:A	NA	frameshift	c.2179_2188delAC TGGCCGGC	p.Thr727fs
2:25240437:C:CC CT	NA	inframe_indel	c.2186_2187insAG G	p.Arg729dup
2:25240437:C:CC GGCCAGTGCCCTC TGAGAGGT	NA	frameshift	c.2174- 9_2186dupACCTCT CAGAGGGCACTGG CCG	p.Leu730fs

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2:25240437:CCG:C	NA	frameshift	c.2185_2186delCG	p.Arg729fs
2:25240438:C:A	NA	missense	c.2186G>T	p.Arg729Leu
2:25240438:C:CG GCCA	NA	frameshift	c.2181_2185dupTG GCC	p.Arg729fs
2:25240438:C:CG GCCAGTGCCTCT GA	NA	frameshift	c.2174- 4_2185dupTCAGA GGGCACTGGCC	p.Arg729fs
2:25240438:C:G	NA	missense	c.2186G>C	p.Arg729Pro
2:25240438:C:T	rs757211277	missense	c.2186G>A	p.Arg729Gln
2:25240438:CG:C	NA	frameshift	c.2185delC	p.Arg729fs
2:25240439:G:A	rs200018028	missense	c.2185C>T	p.Arg729Trp
2:25240439:G:C	rs200018028	missense	c.2185C>G	p.Arg729Gly
2:25240439:GGC CA:G	NA	frameshift	c.2181_2184delTG GC	p.Arg729fs
2:25240440:G:GC	NA	frameshift	c.2183dupG	p.Arg729fs
2:25240440:GC:G	NA	frameshift	c.2183delG	p.Gly728fs
2:25240441:C:A	NA	missense	c.2183G>T	p.Gly728Val
2:25240441:C:CA	NA	frameshift	c.2182_2183insT	p.Gly728fs
2:25240441:C:T	NA	missense	c.2183G>A	p.Gly728Asp
2:25240442:C:A	NA	missense	c.2182G>T	p.Gly728Cys
2:25240442:C:G	NA	missense	c.2182G>C	p.Gly728Arg
2:25240442:C:T	NA	missense	c.2182G>A	p.Gly728Ser
2:25240443:AG:A	NA	frameshift	c.2180delC	p.Thr727fs
2:25240444:GT:G	NA	frameshift	c.2179delA	p.Thr727fs
2:25240445:TG:T	NA	frameshift	c.2178delC	p.Thr727fs
2:25240446:GC:G	NA	frameshift	c.2177delG	p.Gly726fs
2:25240446:GCCC TCTGAGAGGTC:G	NA	frameshift	c.2174- 10_2177delGACCT CTCAGAGGG	p.Glu725fs
2:25240447:C:A	rs1432383727	missense	c.2177G>T	p.Gly726Val
2:25240447:C:T	NA	missense	c.2177G>A	p.Gly726Asp
2:25240448:C:A	NA	missense	c.2176G>T	p.Gly726Cys

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2:25240448:C:G	NA	missense	c.2176G>C	p.Gly726Arg
2:25240448:CCT:C	rs1175429748	frameshift	c.2174_2175delAG	p.Glu725fs
2:25240448:CCTC T:C	NA	frameshift	c.2174- 2_2175delAGAG	p.Glu725fs
2:25240449:CT:C	NA	frameshift	c.2174delA	p.Glu725fs
2:25240451:C:A	NA	splice_acceptor	c.2174-1G>T	
2:25240451:C:G	NA	splice_acceptor	c.2174-1G>C	
2:25240451:C:T	rs1262354256	splice_acceptor	c.2174-1G>A	
2:25240452:T:A	NA	splice_acceptor	c.2174-2A>T	
2:25240452:T:C	rs772010891	splice_acceptor	c.2174-2A>G	
2:25240452:T:G	rs772010891	splice_acceptor	c.2174-2A>C	
2:25240631:GGT ACCTACC:G	NA	frameshift	c.2173_2173+8del GGTAGGTAC	p.Glu725fs
2:25240633:TACC TACCGTA:T	NA	frameshift	c.2170_2173+6delT ACGGTAGGT	p.Tyr724fs
2:25240637:T:TA	NA	splice_donor	c.2173+2dupT	
2:25240638:A:G	NA	splice_donor	c.2173+2T>C	
2:25240638:AC:A	NA	splice_donor	c.2173+1delG	
2:25240639:C:A	NA	splice_donor	c.2173+1G>T	
2:25240639:C:G	NA	splice_donor	c.2173+1G>C	
2:25240639:C:T	rs763716866	splice_donor	c.2173+1G>A	
2:25240640:C:A		stop_gained	c.2173G>T	p.Glu725*
2:25240640:C:CC TA	NA	stop_gained	c.2172_2173insTA G	p.Tyr724_Glu725insTer
2:25240640:C:CG T	NA	frameshift	c.2171_2172dupAC	p.Glu725fs
2:25240641:G:C	NA	stop_gained	c.2172C>G	p.Tyr724*

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2:25240641:G:T	NA	stop_gained	c.2172C>A	p.Tyr724*
2:25240642:T:C	rs1330225644	missense	c.2171A>G	p.Tyr724Cys
2:25240642:T:TCT C	NA	stop_gained	c.2170_2171insGA G	p.Tyr724delinsTer Asp
2:25240644:G:GA	NA	frameshift	c.2168dupT	p.Tyr724fs
2:25240644:GA:G	NA	frameshift	c.2168delT	p.Leu723fs
2:25240645:A:T	NA	missense	c.2168T>A	p.Leu723His
2:25240645:AGG: A	NA	frameshift	c.2166_2167delCC	p.Tyr724fs
2:25240646:G:T	NA	missense	c.2167C>A	p.Leu723Ile
2:25240647:GC:G	NA	frameshift	c.2165delG	p.Gly722fs
2:25240647:GCC: G	NA	frameshift	c.2164_2165delGG	p.Gly722fs
2:25240648:C:T	NA	missense	c.2165G>A	p.Gly722Asp
2:25240650:C:CTT	NA	frameshift	c.2161_2162dupAA	p.Gly722fs
2:25240651:T:A	NA	missense	c.2162A>T	p.Lys721Met
2:25240652:T:A	NA	stop_gained	c.2161A>T	p.Lys721*
2:25240652:T:C	NA	missense	c.2161A>G	p.Lys721Glu
2:25240652:T:TCC GA	NA	frameshift	c.2160_2161insTC GG	p.Lys721fs
2:25240653:GC:G	NA	frameshift	c.2159delG	p.Arg720fs
2:25240654:C:A	rs1272076221	missense	c.2159G>T	p.Arg720Leu
2:25240654:C:G	NA	missense	c.2159G>C	p.Arg720Pro
2:25240654:C:T	NA	missense	c.2159G>A	p.Arg720His
2:25240654:CG:C	NA	frameshift	c.2158delC	p.Arg720fs
2:25240654:CGA GCAGG:C	NA	frameshift	c.2152_2158delCC TGCTC	p.Pro718fs
2:25240655:G:A	rs1197133406	missense	c.2158C>T	p.Arg720Cys
2:25240655:G:C	NA	missense	c.2158C>G	p.Arg720Gly
2:25240655:G:T	NA	missense	c.2158C>A	p.Arg720Ser
2:25240657:G:GT	NA	frameshift	c.2155_2156insA	p.Ala719fs
2:25240657:GCA GGGTT:G	NA	frameshift	c.2149_2155delAA CCCTG	p.Asn717fs

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2:25240658:C:G	NA	missense	c.2155G>C	p.Ala719Pro
2:25240658:C:T	NA	missense	c.2155G>A	p.Ala719Thr
2:25240658:CAG GGTTGACGATGG AG:C	NA	frameshift	c.2139_2154delCT CCATCGTCAACCCT	p.Ser714fs
2:25240659:AG:A	NA	frameshift	c.2153delC	p.Pro718fs
2:25240660:G:A	NA	missense	c.2153C>T	p.Pro718Leu
2:25240660:G:GG GTTGACGAT	NA	frameshift	c.2143_2152dupAT CGTCAACC	p.Pro718fs
2:25240661:G:A	NA	missense	c.2152C>T	p.Pro718Ser
2:25240662:G:GT	NA	frameshift	c.2150dupA	p.Asn717fs
2:25240662:G:GT T	NA	frameshift	c.2149_2150dupAA	p.Asn717fs
2:25240662:GT:G	NA	frameshift	c.2150delA	p.Asn717fs
2:25240663:T:A	NA	missense	c.2150A>T	p.Asn717Ile
2:25240663:T:C	NA	missense	c.2150A>G	p.Asn717Ser
2:25240663:T:TT G	NA	frameshift	c.2148_2149dupCA	p.Asn717fs
2:25240664:TGA: T	NA	frameshift	c.2147_2148delTC	p.Val716fs
2:25240666:A:C	NA	missense	c.2147T>G	p.Val716Gly
2:25240666:A:T	rs778857953	missense	c.2147T>A	p.Val716Asp
2:25240666:AC:A	NA	frameshift	c.2146delG	p.Val716fs
2:25240667:C:A	rs1418039680	missense	c.2146G>T	p.Val716Phe
2:25240667:C:CA	NA	frameshift	c.2145_2146insT	p.Val716fs
2:25240667:C:T	rs1418039680	missense	c.2146G>A	p.Val716Ile
2:25240667:CG:C	NA	frameshift	c.2145delC	p.Ile715fs
2:25240668:GAT: G	NA	frameshift	c.2143_2144delAT	p.Ile715fs
2:25240668:GAT GGAGAGGTC:G	NA	frameshift	c.2134_2144delGA CCTCTCCAT	p.Asp712fs
2:25240669:ATG GAGAGG:A	NA	frameshift	c.2136_2143delCC TCTCCA	p.Leu713fs
2:25240670:T:TG	rs1179539918	frameshift	c.2142dupC	p.Ile715fs

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2:25240670:TG:T	NA	frameshift	c.2142delC	p.Ile715fs
2:25240670:TGG AGA:T	NA	frameshift	c.2138_2142delTCT CC	p.Leu713fs
2:25240671:G:GA	NA	frameshift	c.2141_2142insT	p.Ile715fs
2:25240671:G:GG A	NA	frameshift	c.2140_2141dupTC	p.Ile715fs
2:25240671:G:GG AGA	NA	frameshift	c.2138_2141dupTC TC	p.Ile715fs
2:25240671:GGA: G	rs1281903798	frameshift	c.2140_2141delTC	p.Ser714fs
2:25240672:G:A	NA	missense	c.2141C>T	p.Ser714Phe
2:25240672:G:C	rs367909007	missense	c.2141C>G	p.Ser714Cys
2:25240672:G:GA	NA	frameshift	c.2140dupT	p.Ser714fs
2:25240672:G:T	NA	missense	c.2141C>A	p.Ser714Tyr
2:25240674:GAG GTC:G	NA	frameshift	c.2134_2138delGA CCT	p.Asp712fs
2:25240675:A:AT	NA	frameshift	c.2137_2138insA	p.Leu713fs
2:25240675:AG:A	rs777214520	frameshift	c.2137delC	p.Leu713fs
2:25240675:AGG TCATTGCAG:A	NA	frameshift	c.2127_2137delCT GCAATGACC	p.Cys710fs
2:25240676:G:A	rs780006700	missense	c.2137C>T	p.Leu713Phe
2:25240676:G:C	NA	missense	c.2137C>G	p.Leu713Val
2:25240676:GGT: G	NA	frameshift	c.2135_2136delAC	p.Asp712fs
2:25240677:G:GT	NA	frameshift	c.2135dupA	p.Asp712fs
2:25240677:G:GT CAT	NA	frameshift	c.2132_2135dupAT GA	p.Asp712fs
2:25240677:G:T	NA	missense	c.2136C>A	p.Asp712Glu
2:25240677:GT:G	NA	frameshift	c.2135delA	p.Asp712fs
2:25240678:T:A	NA	missense	c.2135A>T	p.Asp712Val
2:25240678:T:C	NA	missense	c.2135A>G	p.Asp712Gly
2:25240678:T:G	NA	missense	c.2135A>C	p.Asp712Ala
2:25240678:TC:T	NA	frameshift	c.2134delG	p.Asp712fs

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2:25240678:TCAT TGCAGGGACTGC CC:T	NA	frameshift	c.2118_2134delGG GCAGTCCCTGCAAT G	p.Gly707fs
2:25240680:A:AT	NA	frameshift	c.2132dupA	p.Asn711fs
2:25240682:TG:T	NA	frameshift	c.2130delC	p.Cys710fs
2:25240683:G:T	NA	stop_gained	c.2130C>A	p.Cys710*
2:25240683:GCA GGGACTGCCCC:G	NA	frameshift	c.2117_2129delGG GGCAGTCCCTG	p.Gly706fs
2:25240684:C:G	rs1444323322	missense	c.2129G>C	p.Cys710Ser
2:25240684:C:T	NA	missense	c.2129G>A	p.Cys710Tyr
2:25240684:CA:C	NA	frameshift	c.2128delT	p.Cys710fs
2:25240685:A:G	NA	missense	c.2128T>C	p.Cys710Arg
2:25240685:A:T	rs746857175	missense	c.2128T>A	p.Cys710Ser
2:25240685:AG:A	NA	frameshift	c.2127delC	p.Cys710fs
2:25240687:G:A	NA	missense	c.2126C>T	p.Pro709Leu
2:25240687:G:GA	NA	frameshift	c.2125_2126insT	p.Pro709fs
2:25240687:GGA CTGCCCCCAATC:G	NA	frameshift	c.2112_2125delGA TTGGGGGCAGTC	p.Ile705fs
2:25240688:GA:G	NA	frameshift	c.2124delT	p.Cys710fs
2:25240689:AC:A	NA	frameshift	c.2123delG	p.Ser708fs
2:25240689:ACTG CCCCCAATCACCA GATCGAATGGGC CC:A	NA	inframe_indel	c.2094_2123delGG GCCCCATTCGATCTG GTGATTGGGGGCA G	p.Trp698_Ser708 delinsCys
2:25240690:C:A	NA	missense	c.2123G>T	p.Ser708Ile
2:25240690:C:T	NA	missense	c.2123G>A	p.Ser708Asn
2:25240690:CT:C	NA	frameshift	c.2122delA	p.Ser708fs
2:25240690:CTG: C	NA	frameshift	c.2121_2122delCA	p.Pro709fs
2:25240691:TG:T	NA	frameshift	c.2121delC	p.Ser708fs
2:25240692:G:GC	rs1402117804	frameshift	c.2120dupG	p.Ser708fs
2:25240692:GC:G	rs762350913	frameshift	c.2120delG	p.Gly707fs
2:25240693:C:A	NA	missense	c.2120G>T	p.Gly707Val
2:25240693:C:T	rs927337009	missense	c.2120G>A	p.Gly707Asp

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2:25240694:C:A	NA	missense	c.2119G>T	p.Gly707Cys
2:25240694:C:G	NA	missense	c.2119G>C	p.Gly707Arg
2:25240694:C:T	NA	missense	c.2119G>A	p.Gly707Ser
2:25240695:CCCA ATCACCAGATCGA ATGGGCCCCACT: C	NA	frameshift	c.2090_2117delAG TGGGGCCCCATTCTGA TCTGGTGATTGG	p.Glu697fs
2:25240696:C:A	NA	missense	c.2117G>T	p.Gly706Val
2:25240696:C:T	rs777898555	missense	c.2117G>A	p.Gly706Glu
2:25240697:C:A	rs749365376	missense	c.2116G>T	p.Gly706Trp
2:25240697:C:G	NA	missense	c.2116G>C	p.Gly706Arg
2:25240697:C:T	NA	missense	c.2116G>A	p.Gly706Arg
2:25240697:CA:C	NA	frameshift	c.2115delT	p.Ile705fs
2:25240698:AATC ACCAG:A	NA	frameshift	c.2107_2114delCT GGTGAT	p.Leu703fs
2:25240699:A:C	rs777037011	missense	c.2114T>G	p.Ile705Ser
2:25240699:A:G	rs777037011	missense	c.2114T>C	p.Ile705Thr
2:25240699:A:T	NA	missense	c.2114T>A	p.Ile705Asn
2:25240699:AT:A	rs1411778215	frameshift	c.2113delA	p.Ile705fs
2:25240700:T:A	rs1291851173	missense	c.2113A>T	p.Ile705Phe
2:25240700:T:C	NA	missense	c.2113A>G	p.Ile705Val
2:25240700:T:TC A	NA	frameshift	c.2111_2112dupTG	p.Ile705fs
2:25240700:TC:T	NA	frameshift	c.2112delG	p.Ile705fs
2:25240700:TCA: T	NA	frameshift	c.2111_2112delTG	p.Val704fs
2:25240701:CA:C	NA	frameshift	c.2111delT	p.Val704fs
2:25240702:A:C	NA	missense	c.2111T>G	p.Val704Gly
2:25240702:A:G	rs1165074698	missense	c.2111T>C	p.Val704Ala
2:25240702:A:T	NA	missense	c.2111T>A	p.Val704Glu
2:25240702:AC:A	rs1454822561	frameshift	c.2110delG	p.Val704fs
2:25240704:CAG: C	NA	frameshift	c.2107_2108delCT	p.Leu703fs

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2:25240705:A:AG	NA	frameshift	c.2107dupC	p.Leu703fs
2:25240705:A:C	NA	missense	c.2108T>G	p.Leu703Arg
2:25240705:A:G	NA	missense	c.2108T>C	p.Leu703Pro
2:25240705:A:T	NA	missense	c.2108T>A	p.Leu703Gln
2:25240705:AG:A	rs765733093	frameshift	c.2107delC	p.Leu703fs
2:25240706:G:C	NA	missense	c.2107C>G	p.Leu703Val
2:25240706:GAT: G	NA	frameshift	c.2105_2106delAT	p.Asp702fs
2:25240707:A:C	NA	missense	c.2106T>G	p.Asp702Glu
2:25240707:A:T	NA	missense	c.2106T>A	p.Asp702Glu
2:25240707:AT:A	NA	frameshift	c.2105delA	p.Asp702fs
2:25240708:T:A	NA	missense	c.2105A>T	p.Asp702Val
2:25240708:T:C	rs1400330086	missense	c.2105A>G	p.Asp702Gly
2:25240708:T:G	NA	missense	c.2105A>C	p.Asp702Ala
2:25240708:TC:T	NA	frameshift	c.2104delG	p.Asp702fs
2:25240708:TCGA ATGGG:T	NA	frameshift	c.2097_2104delCC CATTCG	p.Pro700fs
2:25240709:C:A	rs770938712	missense	c.2104G>T	p.Asp702Tyr
2:25240709:C:G	NA	missense	c.2104G>C	p.Asp702His
2:25240709:C:T	rs770938712	missense	c.2104G>A	p.Asp702Asn
2:25240710:G:C	NA	missense	c.2103C>G	p.Phe701Leu
2:25240710:G:T	NA	missense	c.2103C>A	p.Phe701Leu
2:25240710:GA:G	NA	frameshift	c.2102delT	p.Phe701fs
2:25240711:A:C	NA	missense	c.2102T>G	p.Phe701Cys
2:25240711:A:G	NA	missense	c.2102T>C	p.Phe701Ser
2:25240711:A:T	NA	missense	c.2102T>A	p.Phe701Tyr
2:25240712:A:C	rs760042734	missense	c.2101T>G	p.Phe701Val
2:25240712:A:G	NA	missense	c.2101T>C	p.Phe701Leu
2:25240712:AT:A	NA	frameshift	c.2100delA	p.Phe701fs
2:25240712:ATG GGCCCCACTCC:A	NA	frameshift	c.2088_2100delGG AGTGGGGCCCA	p.Gln696fs
2:25240713:TG:T	rs773340419	frameshift	c.2099delC	p.Pro700fs
2:25240714:G:A	rs772368909	missense	c.2099C>T	p.Pro700Leu

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2:25240714:G:C	NA	missense	c.2099C>G	p.Pro700Arg
2:25240714:G:T	NA	missense	c.2099C>A	p.Pro700Gln
2:25240715:G:A	rs775823075	missense	c.2098C>T	p.Pro700Ser
2:25240715:G:C	rs775823075	missense	c.2098C>G	p.Pro700Ala
2:25240715:G:T	rs775823075	missense	c.2098C>A	p.Pro700Thr
2:25240716:G:GA	NA	frameshift	c.2096_2097insT	p.Phe701fs
2:25240716:G:GC	rs762965546	frameshift	c.2096dupG	p.Phe701fs
2:25240716:G:GC C	NA	frameshift	c.2095_2096dupG G	p.Pro700fs
2:25240716:G:GC CCCA	NA	frameshift	c.2092_2096dupTG GGG	p.Pro700fs
2:25240716:G:GC CCCCT	NA	frameshift	c.2090_2096dupA GTGGGG	p.Pro700fs
2:25240716:GC:G		frameshift	c.2096delG	p.Gly699fs
2:25240716:GCCC CA:G	NA	frameshift	c.2092_2096delTG GGG	p.Trp698fs
2:25240716:GCCC CACT:G	NA	frameshift	c.2090_2096delAG TGGGG	p.Glu697fs
2:25240717:C:A	NA	missense	c.2096G>T	p.Gly699Val
2:25240717:C:T	rs761064473	missense	c.2096G>A	p.Gly699Asp
2:25240718:C:A	NA	missense	c.2095G>T	p.Gly699Cys
2:25240718:C:G	NA	missense	c.2095G>C	p.Gly699Arg
2:25240718:C:T	rs763776241	missense	c.2095G>A	p.Gly699Ser
2:25240718:CCCA CT:C	NA	frameshift	c.2090_2094delAG TGG	p.Glu697fs
2:25240719:C:A	NA	missense	c.2094G>T	p.Trp698Cys
2:25240719:C:T	rs753452266	stop_gained	c.2094G>A	p.Trp698*
2:25240720:C:G	NA	missense	c.2093G>C	p.Trp698Ser
2:25240720:C:T	rs1005359774	stop_gained	c.2093G>A	p.Trp698*
2:25240720:CA:C	NA	frameshift	c.2092delT	p.Trp698fs
2:25240720:CACT :C	NA	inframe_indel	c.2090_2092delAG T	p.Glu697_Trp698 delinsGly
2:25240721:A:G	NA	missense	c.2092T>C	p.Trp698Arg

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2:25240721:A:T	NA	missense	c.2092T>A	p.Trp698Arg
2:25240721:AC:A	NA	frameshift	c.2091delG	p.Glu697fs
2:25240723:T:TC	NA	frameshift	c.2089dupG	p.Glu697fs
2:25240723:TCC:T	NA	frameshift	c.2088_2089delGG	p.Glu697fs
2:25240724:C:A	rs761343150	stop_gained	c.2089G>T	p.Glu697*
2:25240726:TG:T	rs766594867	frameshift	c.2086delC	p.Gln696fs
2:25240726:TGG: T	NA	frameshift	c.2085_2086delCC	p.Gln696fs
2:25240726:TGG ATCTGGGAGGAT AAAG:T	NA	frameshift	c.2083- 14_2086delCTTTAT CCTCCCAGATCC	p.Ile695fs
2:25240727:G:A	rs750325978	stop_gained	c.2086C>T	p.Gln696*
2:25240727:G:T	NA	missense	c.2086C>A	p.Gln696Lys
2:25240729:A:T	rs780206885	missense	c.2084T>A	p.Ile695Asn
2:25240731:C:A	NA	splice_acceptor	c.2083-1G>T	
2:25240731:C:G	NA	splice_acceptor	c.2083-1G>C	
2:25240731:C:T	NA	splice_acceptor	c.2083-1G>A	
2:25240732:T:A	NA	splice_acceptor	c.2083-2A>T	
2:25240732:T:C	rs761987159	splice_acceptor	c.2083-2A>G	
2:25241553:TGG ACATACATGCTTC TGTGTGACGCTGC GGAC:T	NA	splice_donor	c.2059_2082+8del GTCCGCAGCGTCAC ACAGAAGCATGTAT GTCC	p.Val687_His694del
2:25241559:T:TA	rs751693865	splice_donor	c.2082+2dupT	
2:25241559:TACA TGCTTCTGTGTG:T	NA	frameshift	c.2070_2082+2del CACACAGAAGCATG T	p.Thr691fs
2:25241560:A:C	rs1196735995	splice_donor	c.2082+2T>G	
2:25241560:A:G	rs1196735995	splice_donor	c.2082+2T>C	

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2:25241560:A:T	NA	splice_donor	c.2082+2T>A	
2:25241561:C:A	NA	splice_donor	c.2082+1G>T	
2:25241561:C:CA	NA	frameshift	c.2082dupT	p.Ile695fs
2:25241561:C:CA TGCT	NA	frameshift	c.2078_2082dupA GCAT	p.Ile695fs
2:25241561:C:G	NA	splice_donor	c.2082+1G>C	
2:25241561:C:T	rs766397390	splice_donor	c.2082+1G>A	
2:25241561:CATG CTTCT:C	NA	frameshift	c.2075_2082delAG AAGCAT	p.Gln692fs
2:25241562:AT:A	NA	frameshift	c.2081delA	p.His694fs
2:25241563:T:C	NA	missense	c.2081A>G	p.His694Arg
2:25241563:T:G	NA	missense	c.2081A>C	p.His694Pro
2:25241563:TG:T	NA	frameshift	c.2080delC	p.His694fs
2:25241564:G:A	NA	missense	c.2080C>T	p.His694Tyr
2:25241564:GCTT CTGT:G	NA	frameshift	c.2073_2079delAC AGAAG	p.Gln692fs
2:25241565:C:CTT A	NA	inframe_indel	c.2078_2079insTA A	p.Gln692_Lys693i nsAsn
2:25241565:C:G	NA	missense	c.2079G>C	p.Lys693Asn
2:25241565:CT:C	NA	frameshift	c.2078delA	p.Lys693fs
2:25241566:T:TTC	NA	frameshift	c.2076_2077dupG A	p.Lys693fs
2:25241566:TTC:T	NA	frameshift	c.2076_2077delGA	p.Lys693fs
2:25241567:T:A	rs1365278000	stop_gained	c.2077A>T	p.Lys693*
2:25241568:C:CT	NA	frameshift	c.2075dupA	p.Lys693fs
2:25241568:CTG: C	NA	frameshift	c.2074_2075delCA	p.Gln692fs
2:25241569:TGTG TGACGCTGC:T	NA	inframe_indel	c.2063_2074delGC AGCGTCACAC	p.Arg688_Thr691 del
2:25241570:G:A	rs1278018793	stop_gained	c.2074C>T	p.Gln692*
2:25241570:G:GT	NA	frameshift	c.2073dupA	p.Gln692fs
2:25241570:GT:G	NA	frameshift	c.2073delA	p.Gln692fs
2:25241571:T:TG TG	NA	inframe_indel	c.2070_2072dupCA C	p.Thr691dup

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2:25241571:TG:T	NA	frameshift	c.2072delC	p.Thr691fs
2:25241572:G:A	rs1193025338	missense	c.2072C>T	p.Thr691Ile
2:25241572:G:GT	NA	frameshift	c.2071dupA	p.Thr691fs
2:25241572:GT:G	NA	frameshift	c.2071delA	p.Thr691fs
2:25241573:T:G	NA	missense	c.2071A>C	p.Thr691Pro
2:25241573:T:TA	NA	frameshift	c.2070_2071insT	p.Thr691fs
2:25241574:GA:G	NA	frameshift	c.2069delT	p.Val690fs
2:25241575:A:AC GCTG	NA	frameshift	c.2064_2068dupCA GCG	p.Val690fs
2:25241575:A:C	NA	missense	c.2069T>G	p.Val690Gly
2:25241575:A:G	NA	missense	c.2069T>C	p.Val690Ala
2:25241575:A:T	NA	missense	c.2069T>A	p.Val690Asp
2:25241575:AC:A	NA	frameshift	c.2068delG	p.Val690fs
2:25241576:C:A	NA	missense	c.2068G>T	p.Val690Phe
2:25241576:C:T	rs768899154	missense	c.2068G>A	p.Val690Ile
2:25241576:CG:C	NA	frameshift	c.2067delC	p.Ser689fs
2:25241577:G:GC TGCGGACGTCCCC GACGTACATGAT	NA	frameshift	c.2041_2066dupAT CATGTACGTCTGGGG ACGTCCGCAG	p.Ser689fs
2:25241579:TG:T	NA	frameshift	c.2064delC	p.Ser689fs
2:25241580:GC:G	NA	frameshift	c.2063delG	p.Arg688fs
2:25241580:GCG GACGTCCC:G	NA	frameshift	c.2054_2063delGG GACGTCCG	p.Gly685fs
2:25241581:C:A	NA	missense	c.2063G>T	p.Arg688Leu
2:25241581:C:G	NA	missense	c.2063G>C	p.Arg688Pro
2:25241581:C:T	rs369713081	missense	c.2063G>A	p.Arg688His
2:25241581:CG:C	NA	frameshift	c.2062delC	p.Arg688fs
2:25241582:G:A	rs1484795800	missense	c.2062C>T	p.Arg688Cys
2:25241582:G:C	rs1484795800	missense	c.2062C>G	p.Arg688Gly
2:25241582:G:T	NA	missense	c.2062C>A	p.Arg688Ser
2:25241584:A:C	NA	missense	c.2060T>G	p.Val687Gly
2:25241584:A:G	NA	missense	c.2060T>C	p.Val687Ala
2:25241584:A:T	NA	missense	c.2060T>A	p.Val687Asp

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2:25241585:C:A	NA	missense	c.2059G>T	p.Val687Phe
2:25241585:C:G	NA	missense	c.2059G>C	p.Val687Leu
2:25241586:G:C	NA	missense	c.2058C>G	p.Asp686Glu
2:25241586:G:GT CCCCGACGTACAT GATCA	NA	frameshift	c.2057_2058insTG ATCATGTACGTCGG GGA	p.Val687fs
2:25241586:GT:G	NA	frameshift	c.2057delA	p.Asp686fs
2:25241587:T:A	rs754764419	missense	c.2057A>T	p.Asp686Val
2:25241587:T:C	rs754764419	missense	c.2057A>G	p.Asp686Gly
2:25241587:T:G	NA	missense	c.2057A>C	p.Asp686Ala
2:25241587:T:TC	NA	frameshift	c.2056dupG	p.Asp686fs
2:25241587:TC:T	NA	frameshift	c.2056delG	p.Asp686fs
2:25241587:TCC:T	rs1344775606	frameshift	c.2055_2056delGG	p.Asp686fs
2:25241587:TCCC CGACGTAC:T	NA	frameshift	c.2046_2056delGT ACGTCGGGG	p.Met682fs
2:25241588:C:A	rs767224028	missense	c.2056G>T	p.Asp686Tyr
2:25241588:C:G	NA	missense	c.2056G>C	p.Asp686His
2:25241588:C:T	NA	missense	c.2056G>A	p.Asp686Asn
2:25241589:CCCG ACGTACATGATCT T:C	NA	frameshift	c.2038_2054delAA GATCATGTACGTCG G	p.Lys680fs
2:25241590:C:A	NA	missense	c.2054G>T	p.Gly685Val
2:25241590:C:G	rs754037033	missense	c.2054G>C	p.Gly685Ala
2:25241590:C:T	rs754037033	missense	c.2054G>A	p.Gly685Glu
2:25241590:CCGA CG:C	NA	frameshift	c.2049_2053delCG TCG	p.Tyr683fs
2:25241591:C:A	rs779176507	missense	c.2053G>T	p.Gly685Trp
2:25241591:C:G	NA	missense	c.2053G>C	p.Gly685Arg
2:25241591:C:T	rs779176507	missense	c.2053G>A	p.Gly685Arg
2:25241591:CG:C	NA	frameshift	c.2052delC	p.Asp686fs
2:25241591:CGAC G:C	NA	frameshift	c.2049_2052delCG TC	p.Tyr683fs
2:25241591:CGAC GT:C	NA	frameshift	c.2048_2052delAC GTC	p.Tyr683fs

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2:25241592:GA:G	NA	frameshift	c.2051delT	p.Val684fs
2:25241592:GAC GTAC:G	NA	inframe_indel	c.2046_2051delGT ACGT	p.Met682_Val684 delinsIle
2:25241593:A:C	NA	missense	c.2051T>G	p.Val684Gly
2:25241593:A:T	NA	missense	c.2051T>A	p.Val684Asp
2:25241593:ACGT ACATGATCTTCCC: A	NA	frameshift	c.2035_2050delGG GAAGATCATGTACG	p.Gly679fs
2:25241594:C:A	NA	missense	c.2050G>T	p.Val684Phe
2:25241594:CG:C	NA	frameshift	c.2049delC	p.Tyr683fs
2:25241594:CGTA CATG:C	NA	frameshift	c.2043_2049delCA TGTAC	p.Ile681fs
2:25241595:G:C	NA	stop_gained	c.2049C>G	p.Tyr683*
2:25241595:G:T	NA	stop_gained	c.2049C>A	p.Tyr683*
2:25241595:GT:G	NA	frameshift	c.2048delA	p.Tyr683fs
2:25241596:T:C	rs780495518	missense	c.2048A>G	p.Tyr683Cys
2:25241596:T:TA CATGATCTTCCCC TGGTGCCGC	NA	frameshift	c.2025_2047dupGC GGCACCAGGGGAA GATCATGT	p.Tyr683fs
2:25241596:TACA TG:T	NA	frameshift	c.2043_2047delCA TGT	p.Met682fs
2:25241597:A:C	NA	missense	c.2047T>G	p.Tyr683Asp
2:25241597:A:G	NA	missense	c.2047T>C	p.Tyr683His
2:25241597:A:T	NA	missense	c.2047T>A	p.Tyr683Asn
2:25241598:CA:C	NA	frameshift	c.2045delT	p.Met682fs
2:25241598:CAT: C	NA	frameshift	c.2044_2045delAT	p.Met682fs
2:25241598:CATG :C	NA	inframe_indel	c.2043_2045delCA T	p.Ile681del
2:25241599:A:AT	NA	frameshift	c.2044dupA	p.Met682fs
2:25241599:A:C	rs1324879320	missense	c.2045T>G	p.Met682Arg
2:25241599:ATGA TCTTCCCCTGGT:A	NA	inframe_indel	c.2030_2044delAC CAGGGGAAGATCA	p.His677_Met682 delinsLeu
2:25241600:TG:T	rs1455730245	frameshift	c.2043delC	p.Met682fs

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2:25241600:TGAT CTTCCCCTGGTGC CGCA:T	NA	frameshift	c.2024_2043delTG CGGCACCAGGGGA AGATC	p.Val675fs
2:25241601:G:C		missense	c.2043C>G	p.Ile681Met
2:25241601:G:GT	NA	frameshift	c.2042_2043insA	p.Met682fs
2:25241602:A:AT CTTCCCCTGGTGC CGCACCATGCC	NA	frameshift	c.2017_2041dupG GCATGGTGC GGCA CCAGGGGAAGA	p.Ile681fs
2:25241602:A:G	NA	missense	c.2042T>C	p.Ile681Thr
2:25241602:A:T	rs769006337	missense	c.2042T>A	p.Ile681Asn
2:25241603:T:A	NA	missense	c.2041A>T	p.Ile681Phe
2:25241604:C:A	NA	missense	c.2040G>T	p.Lys680Asn
2:25241604:CT:C	NA	frameshift	c.2039delA	p.Lys680fs
2:25241605:T:C	NA	missense	c.2039A>G	p.Lys680Arg
2:25241606:T:A	NA	stop_gained	c.2038A>T	p.Lys680*
2:25241606:TC:T	rs1365479526	frameshift	c.2037delG	p.Lys680fs
2:25241608:C:A	NA	missense	c.2036G>T	p.Gly679Val
2:25241608:C:T	NA	missense	c.2036G>A	p.Gly679Glu
2:25241609:C:A	NA	missense	c.2035G>T	p.Gly679Trp
2:25241610:CT:C	NA	frameshift	c.2033delA	p.Gln678fs
2:25241611:T:TG	NA	frameshift	c.2032dupC	p.Gln678fs
2:25241611:TG:T	NA	frameshift	c.2032delC	p.Gln678fs
2:25241612:G:A	rs1050271318	stop_gained	c.2032C>T	p.Gln678*
2:25241614:T:C	NA	missense	c.2030A>G	p.His677Arg
2:25241614:T:TG CCG	NA	frameshift	c.2026_2029dupCG GC	p.His677fs
2:25241615:GCC GCA:G	NA	frameshift	c.2024_2028delTG CGG	p.Val675fs
2:25241616:C:CC G	NA	frameshift	c.2026_2027dupCG	p.His677fs
2:25241617:C:A	rs769539104	missense	c.2027G>T	p.Arg676Leu
2:25241617:C:T	rs769539104	missense	c.2027G>A	p.Arg676Gln
2:25241617:CG:C	NA	frameshift	c.2026delC	p.Arg676fs

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2:25241618:G:A	rs375399431	missense	c.2026C>T	p.Arg676Trp
2:25241618:G:C	NA	missense	c.2026C>G	p.Arg676Gly
2:25241618:G:GC	NA	frameshift	c.2025dupG	p.Arg676fs
2:25241618:G:GC ACCA	NA	frameshift	c.2021_2025dupTG GTG	p.Arg676fs
2:25241618:GCAC CATGCCACCGTG ATGGAGTCCT:G	NA	frameshift	c.2000_2025delAG GACTCCATCACGGT GGGCATGGTG	p.Glu667fs
2:25241619:CA:C	NA	frameshift	c.2024delT	p.Val675fs
2:25241620:A:AC	NA	frameshift	c.2023dupG	p.Val675fs
2:25241620:A:C		missense	c.2024T>G	p.Val675Gly
2:25241620:A:G		missense	c.2024T>C	p.Val675Ala
2:25241620:A:T	NA	missense	c.2024T>A	p.Val675Glu
2:25241620:AC:A	rs763843749	frameshift	c.2023delG	p.Val675fs
2:25241621:C:A	NA	missense	c.2023G>T	p.Val675Leu
2:25241621:C:G	NA	missense	c.2023G>C	p.Val675Leu
2:25241621:C:T	rs1025238838	missense	c.2023G>A	p.Val675Met
2:25241623:AT:A	NA	frameshift	c.2020delA	p.Met674fs
2:25241623:ATGC CCAC:A	NA	frameshift	c.2014_2020delGT GGGCA	p.Val672fs
2:25241624:T:C	rs1306265898	missense	c.2020A>G	p.Met674Val
2:25241624:TG:T	NA	frameshift	c.2019delC	p.Met674fs
2:25241625:G:GC C	NA	frameshift	c.2017_2018dupG G	p.Met674fs
2:25241625:G:GC CC	NA	inframe_indel	c.2016_2018dupG GG	p.Gly673dup
2:25241625:G:GC CCA	NA	frameshift	c.2015_2018dupTG GG	p.Met674fs
2:25241625:GC:G	NA	frameshift	c.2018delG	p.Gly673fs
2:25241625:GCCC A:G	NA	frameshift	c.2015_2018delTG GG	p.Val672fs
2:25241626:C:A	NA	missense	c.2018G>T	p.Gly673Val
2:25241626:C:T	rs902730598	missense	c.2018G>A	p.Gly673Asp
2:25241627:C:A	NA	missense	c.2017G>T	p.Gly673Cys

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2:25241629:AC:A	NA	frameshift	c.2014delG	p.Val672fs
2:25241630:C:T	NA	missense	c.2014G>A	p.Val672Met
2:25241630:CCGT GATGGAGTCCT:C	NA	frameshift	c.2000_2013delAG GACTCCATCACG	p.Glu667fs
2:25241631:CGTG ATGGA:C	NA	frameshift	c.2005_2012delTC CATCAC	p.Ser669fs
2:25241632:G:A	NA	missense	c.2012C>T	p.Thr671Met
2:25241632:G:GT	NA	frameshift	c.2011dupA	p.Thr671fs
2:25241632:GT:G	NA	frameshift	c.2011delA	p.Thr671fs
2:25241633:TGAT G:T	NA	frameshift	c.2007_2010delCA TC	p.Ile670fs
2:25241634:GAT: G	NA	frameshift	c.2008_2009delAT	p.Ile670fs
2:25241634:GAT GGAGTCCTCACAC A:G	NA	frameshift	c.1994_2009delTG TGTGAGGACTCCAT	p.Val665fs
2:25241635:A:AG GACTCCATCACGG	NA	frameshift	c.2008_2009insCC GTGATGGAGTCC	p.Ile670fs
2:25241635:A:AT GGAGTCCTCACAC ACCTCCGAGGCAA TGTAGCGG	NA	frameshift	c.1974_2008dupCC GCTACATTGCCTCG GAGGTGTGTGAGG ACTCCA	p.Ile670fs
2:25241635:AT:A	NA	frameshift	c.2008delA	p.Ile670fs
2:25241635:ATG GAG:A	NA	frameshift	c.2004_2008delCT CCA	p.Ser669fs
2:25241636:T:A	NA	missense	c.2008A>T	p.Ile670Phe
2:25241636:T:G	NA	missense	c.2008A>C	p.Ile670Leu
2:25241636:T:TG	rs753637583	frameshift	c.2007dupC	p.Ile670fs
2:25241636:TG:T	NA	frameshift	c.2007delC	p.Ile670fs
2:25241637:GGA: G	NA	frameshift	c.2005_2006delTC	p.Ser669fs
2:25241637:GGA GTCCTCA:G	NA	inframe_indel	c.1998_2006delTG AGGACTC	p.Glu667_Ser669 del
2:25241638:G:A	NA	missense	c.2006C>T	p.Ser669Phe

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2:25241638:G:T	NA	missense	c.2006C>A	p.Ser669Tyr
2:25241639:AG:A	NA	frameshift	c.2004delC	p.Ser669fs
2:25241641:T:A	NA	missense	c.2003A>T	p.Asp668Val
2:25241641:T:TC	NA	frameshift	c.2002dupG	p.Asp668fs
2:25241641:TC:T	NA	frameshift	c.2002delG	p.Asp668fs
2:25241642:C:T	NA	missense	c.2002G>A	p.Asp668Asn
2:25241642:CCT: C	NA	frameshift	c.2000_2001delAG	p.Glu667fs
2:25241643:CT:C	NA	frameshift	c.2000delA	p.Glu667fs
2:25241644:T:C	NA	missense	c.2000A>G	p.Glu667Gly
2:25241644:TCA: T	rs1467575781	frameshift	c.1998_1999delTG	p.Cys666fs
2:25241644:TCAC A:T	NA	frameshift	c.1996_1999delTG TG	p.Cys666fs
2:25241645:C:A	NA	stop_gained	c.1999G>T	p.Glu667*
2:25241645:C:CA	NA	frameshift	c.1998dupT	p.Glu667fs
2:25241646:A:C	NA	missense	c.1998T>G	p.Cys666Trp
2:25241646:A:T	NA	stop_gained	c.1998T>A	p.Cys666*
2:25241647:C:A	NA	missense	c.1997G>T	p.Cys666Phe
2:25241647:C:T	NA	missense	c.1997G>A	p.Cys666Tyr
2:25241648:A:C	NA	missense	c.1996T>G	p.Cys666Gly
2:25241649:CA:C	rs1268925454	frameshift	c.1994delT	p.Val665fs
2:25241650:A:C	rs762503226	missense	c.1994T>G	p.Val665Gly
2:25241650:A:G	NA	missense	c.1994T>C	p.Val665Ala
2:25241650:A:T	rs762503226	missense	c.1994T>A	p.Val665Glu
2:25241650:AC:A	NA	frameshift	c.1993delG	p.Val665fs
2:25241651:C:A	rs766020170	missense	c.1993G>T	p.Val665Leu
2:25241651:C:T	NA	missense	c.1993G>A	p.Val665Met
2:25241652:CT:C	NA	frameshift	c.1991delA	p.Glu664fs
2:25241652:CTCC GAGGCAATGTAG CGG:C	NA	inframe_indel	c.1974_1991delCC GCTACATTGCCTCG GA	p.Asp658_Ser663 del
2:25241653:T:C	NA	missense	c.1991A>G	p.Glu664Gly

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2:25241653:TC:T	NA	frameshift	c.1990delG	p.Glu664fs
2:25241653:TCCG AGGCAATGTAG:T	NA	frameshift	c.1977_1990delCT ACATTGCCTCGG	p.Tyr660fs
2:25241654:C:A	rs1476901854	stop_gained	c.1990G>T	p.Glu664*
2:25241654:C:G	NA	missense	c.1990G>C	p.Glu664Gln
2:25241656:G:A	rs553080210	missense	c.1988C>T	p.Ser663Leu
2:25241656:G:C	NA	missense	c.1988C>G	p.Ser663Trp
2:25241656:G:T	NA	stop_gained	c.1988C>A	p.Ser663*
2:25241656:GAG GCAATGT:G	NA	inframe_indel	c.1979_1987delAC ATTGCCT	p.Tyr660_Ala662 del
2:25241657:A:G	NA	missense	c.1987T>C	p.Ser663Pro
2:25241657:AG:A	NA	frameshift	c.1986delC	p.Ser663fs
2:25241658:GGC AATGTA:G	NA	frameshift	c.1978_1985delTA CATTGC	p.Tyr660fs
2:25241659:G:A	NA	missense	c.1985C>T	p.Ala662Val
2:25241659:G:T	NA	missense	c.1985C>A	p.Ala662Asp
2:25241659:GCA AT:G	NA	frameshift	c.1981_1984delAT TG	p.Ile661fs
2:25241660:C:A	NA	missense	c.1984G>T	p.Ala662Ser
2:25241660:C:CA	NA	frameshift	c.1983dupT	p.Ala662fs
2:25241660:C:G	rs759408234	missense	c.1984G>C	p.Ala662Pro
2:25241660:C:T	rs759408234	missense	c.1984G>A	p.Ala662Thr
2:25241660:CAA: C	NA	frameshift	c.1982_1983delTT	p.Ile661fs
2:25241661:A:AA TGTAGCGGTCCAC CTGAATGCCCAAG TCCTTCAGCACCA GGAG	NA	frameshift	c.1939_1982dupCT CCTGGTGCTGAAGG ACTTGGGCATTGAG GTGGACCGCTACAT	p.Ala662fs
2:25241661:AATG T:A	NA	frameshift	c.1979_1982delAC AT	p.Tyr660fs
2:25241662:A:C	NA	missense	c.1982T>G	p.Ile661Ser
2:25241662:A:G	rs1471917721	missense	c.1982T>C	p.Ile661Thr
2:25241662:A:T	NA	missense	c.1982T>A	p.Ile661Asn

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2:25241662:AT:A	NA	frameshift	c.1981delA	p.Ile661fs
2:25241662:ATGT AGC:A	NA	inframe_indel	c.1976_1981delGC TACA	p.Arg659_Ile661del insLeu
2:25241664:G:C	NA	stop_gained	c.1980C>G	p.Tyr660*
2:25241664:G:GT	rs1158856353	frameshift	c.1979dupA	p.Tyr660fs
2:25241665:T:A	NA	missense	c.1979A>T	p.Tyr660Phe
2:25241665:T:C	rs767552800	missense	c.1979A>G	p.Tyr660Cys
2:25241665:T:G	NA	missense	c.1979A>C	p.Tyr660Ser
2:25241666:A:C	NA	missense	c.1978T>G	p.Tyr660Asp
2:25241666:A:G	NA	missense	c.1978T>C	p.Tyr660His
2:25241666:A:T	NA	missense	c.1978T>A	p.Tyr660Asn
2:25241668:C:CG	NA	frameshift	c.1975dupC	p.Arg659fs
2:25241668:C:T	rs752434188	missense	c.1976G>A	p.Arg659His
2:25241668:CG:C	NA	frameshift	c.1975delC	p.Arg659fs
2:25241669:G:A	rs755982635	missense	c.1975C>T	p.Arg659Cys
2:25241669:G:C	rs755982635	missense	c.1975C>G	p.Arg659Gly
2:25241671:T:A	rs146164726	missense	c.1973A>T	p.Asp658Val
2:25241671:T:TC	NA	frameshift	c.1972dupG	p.Asp658fs
2:25241671:T:TCC	NA	frameshift	c.1971_1972dupG G	p.Asp658fs
2:25241671:TC:T	NA	frameshift	c.1972delG	p.Asp658fs
2:25241671:TCCA CCTGAATGC:T	NA	inframe_indel	c.1961_1972delGC ATTCAGGTGG	p.Gly654_Val657 del
2:25241672:C:A	NA	missense	c.1972G>T	p.Asp658Tyr
2:25241672:C:CC A	NA	frameshift	c.1970_1971dupTG	p.Asp658fs
2:25241672:C:T	NA	missense	c.1972G>A	p.Asp658Asn
2:25241672:CCA: C	NA	frameshift	c.1970_1971delTG	p.Val657fs
2:25241672:CCAC CTGAA:C	NA	frameshift	c.1964_1971delTTC AGGTG	p.Ile655fs
2:25241674:A:C	NA	missense	c.1970T>G	p.Val657Gly
2:25241674:A:G	NA	missense	c.1970T>C	p.Val657Ala

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2:25241674:A:T	NA	missense	c.1970T>A	p.Val657Glu
2:25241674:AC:A	NA	frameshift	c.1969delG	p.Val657fs
2:25241674:ACCT GAATG:A	NA	frameshift	c.1962_1969delCA TTCAGG	p.Ile655fs
2:25241674:ACCT GAATGCC:A	NA	frameshift	c.1960_1969delGG CATTCAGG	p.Gly654fs
2:25241675:C:A	NA	missense	c.1969G>T	p.Val657Leu
2:25241675:C:G	NA	missense	c.1969G>C	p.Val657Leu
2:25241675:C:T	rs368961181	missense	c.1969G>A	p.Val657Met
2:25241676:CT:C	NA	frameshift	c.1967delA	p.Gln656fs
2:25241677:T:G	rs1323531460	missense	c.1967A>C	p.Gln656Pro
2:25241677:TG:T	NA	frameshift	c.1966delC	p.Gln656fs
2:25241678:G:A	rs758402405	stop_gained	c.1966C>T	p.Gln656*
2:25241678:GAAT GCCCAAGT:G	NA	frameshift	c.1955_1965delAC TTGGGCATT	p.Asp652fs
2:25241679:A:C	NA	missense	c.1965T>G	p.Ile655Met
2:25241679:AATG C:A	NA	frameshift	c.1961_1964delGC AT	p.Gly654fs
2:25241680:A:C	NA	missense	c.1964T>G	p.Ile655Ser
2:25241680:A:G	NA	missense	c.1964T>C	p.Ile655Thr
2:25241680:A:T	rs780120142	missense	c.1964T>A	p.Ile655Asn
2:25241680:AT:A	NA	frameshift	c.1963delA	p.Ile655fs
2:25241681:T:C	NA	missense	c.1963A>G	p.Ile655Val
2:25241681:T:TG C	NA	frameshift	c.1961_1962dupGC	p.Ile655fs
2:25241681:T:TTT CA	NA	frameshift	c.1962_1963insTG AA	p.Ile655fs
2:25241681:TG:T	NA	frameshift	c.1962delC	p.Ile655fs
2:25241682:G:GC	NA	frameshift	c.1961dupG	p.Ile655fs
2:25241682:GC:G	NA	frameshift	c.1961delG	p.Gly654fs
2:25241682:GCCC AAGTCCTCAGCA :G	NA	frameshift	c.1946_1961delTG CTGAAGGACTTGG G	p.Val649fs
2:25241683:C:A	NA	missense	c.1961G>T	p.Gly654Val

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2:25241683:C:G	NA	missense	c.1961G>C	p.Gly654Ala
2:25241683:C:T	rs1297531346	missense	c.1961G>A	p.Gly654Asp
2:25241684:C:CC T	NA	frameshift	c.1959_1960insAG	p.Gly654fs
2:25241684:C:T	rs1371093470	missense	c.1960G>A	p.Gly654Ser
2:25241685:C:A	rs747168987	missense	c.1959G>T	p.Leu653Phe
2:25241685:C:G	NA	missense	c.1959G>C	p.Leu653Phe
2:25241685:CA:C	NA	frameshift	c.1958delT	p.Leu653fs
2:25241686:A:C	NA	missense	c.1958T>G	p.Leu653Trp
2:25241686:A:G	NA	missense	c.1958T>C	p.Leu653Ser
2:25241686:A:T	rs1202526721	stop_gained	c.1958T>A	p.Leu653*
2:25241686:AAGT CCTTCAG:A	NA	frameshift	c.1948_1957delCT GAAGGACT	p.Leu650fs
2:25241687:A:C	rs1461998130	missense	c.1957T>G	p.Leu653Val
2:25241687:A:T	NA	missense	c.1957T>A	p.Leu653Met
2:25241687:AG:A	NA	frameshift	c.1956delC	p.Leu653fs
2:25241688:G:GT CCT	NA	frameshift	c.1952_1955dupA GGA	p.Asp652fs
2:25241688:G:GT CCTTCA	NA	frameshift	c.1949_1955dupTG AAGGA	p.Leu653fs
2:25241688:GT:G	NA	frameshift	c.1955delA	p.Asp652fs
2:25241689:TC:T	NA	frameshift	c.1954delG	p.Asp652fs
2:25241689:TCCT TCAGCA:T	NA	inframe_indel	c.1946_1954delTG CTGAAGG	p.Val649_Lys651d el
2:25241689:TCCT TCAGCAC:T	NA	frameshift	c.1945_1954delGT GCTGAAGG	p.Val649fs
2:25241691:CT:C	NA	frameshift	c.1952delA	p.Lys651fs
2:25241693:T:A	NA	stop_gained	c.1951A>T	p.Lys651*
2:25241693:TCA: T	NA	frameshift	c.1949_1950delTG	p.Leu650fs
2:25241693:TCAG CA:T	NA	frameshift	c.1946_1950delTG CTG	p.Val649fs
2:25241694:C:CA	NA	frameshift	c.1949dupT	p.Lys651fs
2:25241694:CA:C	NA	frameshift	c.1949delT	p.Leu650fs

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2:25241695:A:C	NA	missense	c.1949T>G	p.Leu650Arg
2:25241695:A:G	NA	missense	c.1949T>C	p.Leu650Pro
2:25241695:A:T	rs1467909574	missense	c.1949T>A	p.Leu650Gln
2:25241695:AGC ACCAGGAG:A	NA	frameshift	c.1939_1948delCT CCTGGTGC	p.Leu647fs
2:25241696:G:C	rs1334392444	missense	c.1948C>G	p.Leu650Val
2:25241696:GC:G	NA	frameshift	c.1947delG	p.Leu650fs
2:25241696:GCA: G	NA	frameshift	c.1946_1947delTG	p.Val649fs
2:25241697:CA:C	NA	frameshift	c.1946delT	p.Val649fs
2:25241698:A:C	NA	missense	c.1946T>G	p.Val649Gly
2:25241698:A:T	NA	missense	c.1946T>A	p.Val649Glu
2:25241699:C:A	NA	missense	c.1945G>T	p.Val649Leu
2:25241699:C:CA	NA	frameshift	c.1944_1945insT	p.Val649fs
2:25241699:C:G	NA	missense	c.1945G>C	p.Val649Leu
2:25241699:C:T	NA	missense	c.1945G>A	p.Val649Met
2:25241700:C:CA GG	NA	inframe_indel	c.1941_1943dupCC T	p.Leu648dup
2:25241700:CA:C	NA	frameshift	c.1943delT	p.Leu648fs
2:25241701:A:C	NA	missense	c.1943T>G	p.Leu648Arg
2:25241701:A:G	rs587777507	missense	c.1943T>C	p.Leu648Pro
2:25241701:AG:A	rs1292349089	frameshift	c.1942delC	p.Leu648fs
2:25241701:AGG AGCCCTGCACCAG C:A	NA	splice_acceptor	c.1937- 10_1942delGCTGG TGCAGGGCTCC	p.Gly646_Leu648 delinsVal
2:25241701:AGG AGCCCTGCACCAG CC:A	NA	splice_acceptor	c.1937- 11_1942delGGCTG GTGCAGGGCTCC	p.Gly646_Leu648 delinsVal
2:25241704:A:AG C	NA	frameshift	c.1938_1939dupGC	p.Leu647fs
2:25241704:A:C	rs781742148	missense	c.1940T>G	p.Leu647Arg
2:25241704:A:G	NA	missense	c.1940T>C	p.Leu647Pro
2:25241704:A:T	rs781742148	missense	c.1940T>A	p.Leu647His
2:25241705:G:C	NA	missense	c.1939C>G	p.Leu647Val

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2:25241705:G:GC	NA	frameshift	c.1938dupG	p.Leu647fs
2:25241705:GC:G	NA	frameshift	c.1938delG	p.Leu647fs
2:25241707:C:T	NA	missense	c.1937G>A	p.Gly646Glu
2:25241708:C:A	NA	splice_acceptor	c.1937-1G>T	
2:25241708:C:G	NA	splice_acceptor	c.1937-1G>C	
2:25241708:C:T	rs748440577	splice_acceptor	c.1937-1G>A	
2:25241708:CT:C	NA	splice_acceptor	c.1937-2delA	
2:25241709:T:A	NA	splice_acceptor	c.1937-2A>T	
2:25241709:T:C	rs770305758	splice_acceptor	c.1937-2A>G	
2:25241709:T:G	NA	splice_acceptor	c.1937-2A>C	
2:25243893:CTCA CCTGTAGCGA:C	NA	splice_donor	c.1928_1936+4delT CGCTACAGGTGA	p.Ile643_Gly646delinsArg
2:25243895:C:CA	NA	splice_donor	c.1936+2dupT	
2:25243896:A:G	rs1190050788	splice_donor	c.1936+2T>C	
2:25243896:A:T	NA	splice_donor	c.1936+2T>A	
2:25243896:AC:A	NA	splice_donor	c.1936+1delG	
2:25243897:C:A	rs1238786275	splice_donor	c.1936+1G>T	
2:25243897:C:G	NA	splice_donor	c.1936+1G>C	
2:25243897:C:T	rs1238786275	splice_donor	c.1936+1G>A	
2:25243898:C:A	rs1472933404	missense	c.1936G>T	p.Gly646Trp
2:25243898:C:G	NA	missense	c.1936G>C	p.Gly646Arg
2:25243898:C:T	NA	missense	c.1936G>A	p.Gly646Arg
2:25243898:CT:C	NA	frameshift	c.1935delA	p.Leu647fs
2:25243900:G:A	rs1387749234	missense	c.1934C>T	p.Thr645Ile
2:25243900:G:GT	NA	frameshift	c.1933dupA	p.Thr645fs
2:25243901:T:C		missense	c.1933A>G	p.Thr645Ala
2:25243903:G:A	NA	missense	c.1931C>T	p.Ala644Val

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2:25243903:GC:G	NA	frameshift	c.1930delG	p.Ala644fs
2:25243904:C:G	NA	missense	c.1930G>C	p.Ala644Pro
2:25243904:C:T	rs1431156021	missense	c.1930G>A	p.Ala644Thr
2:25243904:CG:C	NA	frameshift	c.1929delC	p.Ile643fs
2:25243905:G:GA	NA	frameshift	c.1928dupT	p.Ala644fs
2:25243905:G:GA TTCC	NA	frameshift	c.1924_1928dupG GAAT	p.Ile643fs
2:25243905:GA:G	NA	frameshift	c.1928delT	p.Ile643fs
2:25243905:GATT CCATCAAAGAGA GA:G	NA	frameshift	c.1912_1928delTCT CTCTTTGATGGAAT	p.Ser638fs
2:25243906:A:G	NA	missense	c.1928T>C	p.Ile643Thr
2:25243906:AT:A	NA	frameshift	c.1927delA	p.Ile643fs
2:25243907:T:C	NA	missense	c.1927A>G	p.Ile643Val
2:25243907:T:TTC CA	NA	frameshift	c.1923_1926dupTG GA	p.Ile643fs
2:25243908:T:TC	NA	frameshift	c.1925dupG	p.Ile643fs
2:25243908:TCC:T	NA	frameshift	c.1924_1925delGG	p.Gly642fs
2:25243909:C:A	NA	missense	c.1925G>T	p.Gly642Val
2:25243909:C:CC A	NA	frameshift	c.1923_1924dupTG	p.Gly642fs
2:25243909:C:T	NA	missense	c.1925G>A	p.Gly642Glu
2:25243910:C:A	rs551213321	stop_gained	c.1924G>T	p.Gly642*
2:25243910:C:T	rs551213321	missense	c.1924G>A	p.Gly642Arg
2:25243910:CA:C	NA	frameshift	c.1923delT	p.Asp641fs
2:25243911:A:T	NA	missense	c.1923T>A	p.Asp641Glu
2:25243912:T:A	NA	missense	c.1922A>T	p.Asp641Val
2:25243912:TCAA AGA:T	NA	inframe_indel	c.1916_1921delTCT TTG	p.Leu639_Asp641 delinsHis
2:25243912:TCAA AGAGAGACAG:T	NA	frameshift	c.1909_1921delCT GTCTCTCTTTG	p.Leu637fs
2:25243913:C:A	NA	missense	c.1921G>T	p.Asp641Tyr
2:25243913:C:CA	NA	frameshift	c.1920dupT	p.Asp641fs

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2:25243913:C:G	NA	missense	c.1921G>C	p.Asp641His
2:25243913:C:T	rs1331365627	missense	c.1921G>A	p.Asp641Asn
2:25243913:CA:C	NA	frameshift	c.1920delT	p.Phe640fs
2:25243913:CAA: C	rs1412485785	frameshift	c.1919_1920delTT	p.Phe640fs
2:25243913:CAAA G:C	NA	frameshift	c.1917_1920delCTT T	p.Phe640fs
2:25243913:CAAA GA:C	NA	frameshift	c.1916_1920delTCT TT	p.Leu639fs
2:25243915:A:C	NA	missense	c.1919T>G	p.Phe640Cys
2:25243915:A:G	NA	missense	c.1919T>C	p.Phe640Ser
2:25243915:AAG: A	NA	frameshift	c.1917_1918delCT	p.Phe640fs
2:25243915:AAG AG:A	NA	frameshift	c.1915_1918delCT CT	p.Phe640fs
2:25243915:AAG AGAGAC:A	NA	frameshift	c.1911_1918delGT CTCTCT	p.Ser638fs
2:25243916:A:C	NA	missense	c.1918T>G	p.Phe640Val
2:25243916:A:G	NA	missense	c.1918T>C	p.Phe640Leu
2:25243916:A:T	NA	missense	c.1918T>A	p.Phe640Ile
2:25243917:GA:G	NA	frameshift	c.1916delT	p.Leu639fs
2:25243918:A:C	NA	missense	c.1916T>G	p.Leu639Arg
2:25243918:A:G	NA	missense	c.1916T>C	p.Leu639Pro
2:25243918:A:T	NA	missense	c.1916T>A	p.Leu639His
2:25243918:AG:A	NA	frameshift	c.1915delC	p.Leu639fs
2:25243919:G:A	NA	missense	c.1915C>T	p.Leu639Phe
2:25243919:G:C	NA	missense	c.1915C>G	p.Leu639Val
2:25243919:G:GA GAC	NA	frameshift	c.1911_1914dupGT CT	p.Leu639fs
2:25243919:GA:G	NA	frameshift	c.1914delT	p.Leu639fs
2:25243919:GAG AC:G	NA	frameshift	c.1911_1914delGT CT	p.Phe640fs
2:25243920:A:AG	NA	frameshift	c.1913dupC	p.Leu639fs
2:25243921:G:A	rs1253868717	missense	c.1913C>T	p.Ser638Phe

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2:25243921:G:C	rs1253868717	missense	c.1913C>G	p.Ser638Cys
2:25243921:G:T	NA	missense	c.1913C>A	p.Ser638Tyr
2:25243921:GAC: G	NA	frameshift	c.1911_1912delGT	p.Phe640fs
2:25243922:A:C	NA	missense	c.1912T>G	p.Ser638Ala
2:25243922:A:G	NA	missense	c.1912T>C	p.Ser638Pro
2:25243922:AC:A	NA	frameshift	c.1911delG	p.Ser638fs
2:25243923:CA:C	NA	frameshift	c.1910delT	p.Leu637fs
2:25243924:A:C	rs371523368	missense	c.1910T>G	p.Leu637Arg
2:25243924:A:G	rs371523368	missense	c.1910T>C	p.Leu637Pro
2:25243924:A:T	NA	missense	c.1910T>A	p.Leu637Gln
2:25243925:G:C	NA	missense	c.1909C>G	p.Leu637Val
2:25243925:G:GC A	NA	frameshift	c.1907_1908dupTG	p.Leu637fs
2:25243925:G:GT	NA	frameshift	c.1908_1909insA	p.Leu637fs
2:25243926:CA:C	NA	frameshift	c.1907delT	p.Val636fs
2:25243927:A:AC CCGGAT	NA	frameshift	c.1900_1906dupAT CCGGG	p.Val636fs
2:25243927:A:C	rs929806900	missense	c.1907T>G	p.Val636Gly
2:25243927:A:G	NA	missense	c.1907T>C	p.Val636Ala
2:25243927:A:T	NA	missense	c.1907T>A	p.Val636Glu
2:25243927:AC:A	rs778979256	frameshift	c.1906delG	p.Val636fs
2:25243927:ACCC GG:A	NA	frameshift	c.1902_1906delCC GGG	p.Arg635fs
2:25243928:C:A	rs376550450	missense	c.1906G>T	p.Val636Leu
2:25243928:C:CA	NA	frameshift	c.1905_1906insT	p.Val636fs
2:25243928:C:G	NA	missense	c.1906G>C	p.Val636Leu
2:25243928:C:T	rs376550450	missense	c.1906G>A	p.Val636Met
2:25243930:C:A	NA	missense	c.1904G>T	p.Arg635Leu
2:25243930:C:G	rs751562376	missense	c.1904G>C	p.Arg635Pro
2:25243930:C:T	rs751562376	missense	c.1904G>A	p.Arg635Gln
2:25243930:CG:C	rs1326186258	frameshift	c.1903delC	p.Arg635fs

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2:25243930:CGG ATGGGCTTCCTCT TCTCAGCTGGGAC AG:C	NA	inframe_indel	c.1874_1903delCT GTCCCAGCTGAGAA GAGGAAGCCCATCC	p.Pro625_Ile634del
2:25243931:G:A	rs144689354	missense	c.1903C>T	p.Arg635Trp
2:25243931:G:C	NA	missense	c.1903C>G	p.Arg635Gly
2:25243932:G:C	NA	missense	c.1902C>G	p.Ile634Met
2:25243932:G:GA TGGGCTT	NA	frameshift	c.1894_1901dupAA GCCCCAT	p.Arg635fs
2:25243932:GA:G	NA	frameshift	c.1901delT	p.Ile634fs
2:25243933:A:AT G	NA	frameshift	c.1899_1900dupCA	p.Ile634fs
2:25243933:A:C	NA	missense	c.1901T>G	p.Ile634Ser
2:25243933:A:G	rs1390273539	missense	c.1901T>C	p.Ile634Thr
2:25243933:A:T	NA	missense	c.1901T>A	p.Ile634Asn
2:25243933:AT:A	NA	frameshift	c.1900delA	p.Ile634fs
2:25243934:T:A	NA	missense	c.1900A>T	p.Ile634Phe
2:25243934:TG:T	NA	frameshift	c.1899delC	p.Ile634fs
2:25243936:G:A	rs1446825883	missense	c.1898C>T	p.Pro633Leu
2:25243936:G:C	rs1446825883	missense	c.1898C>G	p.Pro633Arg
2:25243936:G:GG CTT	NA	frameshift	c.1894_1897dupAA GC	p.Pro633fs
2:25243936:G:T	NA	missense	c.1898C>A	p.Pro633His
2:25243937:G:A	rs1191994998	missense	c.1897C>T	p.Pro633Ser
2:25243937:G:GC	NA	frameshift	c.1896dupG	p.Pro633fs
2:25243937:G:T	NA	missense	c.1897C>A	p.Pro633Thr
2:25243937:GC:G	NA	frameshift	c.1896delG	p.Lys632fs
2:25243938:CT:C	rs1195836932	frameshift	c.1895delA	p.Lys632fs
2:25243939:T:TTC CTCTTCTCAGCTG GGACA	NA	frameshift	c.1875_1894dupTG TCCCAGCTGAGAAG AGGA	p.Lys632fs
2:25243940:T:A	rs1245875087	stop_gained	c.1894A>T	p.Lys632*
2:25243940:T:G	NA	missense	c.1894A>C	p.Lys632Gln
2:25243940:TC:T	NA	frameshift	c.1893delG	p.Lys632fs

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2:25243941:C:A	NA	missense	c.1893G>T	p.Arg631Ser
2:25243941:C:G	NA	missense	c.1893G>C	p.Arg631Ser
2:25243941:CCTC TTCTCAGCTG:C	NA	frameshift	c.1880_1892delCA GCTGAGAAGAG	p.Pro627fs
2:25243942:C:A	NA	missense	c.1892G>T	p.Arg631Met
2:25243942:C:G	rs1159270298	missense	c.1892G>C	p.Arg631Thr
2:25243942:C:T	NA	missense	c.1892G>A	p.Arg631Lys
2:25243943:T:A	rs1450721986	missense	c.1891A>T	p.Arg631Trp
2:25243943:T:C	NA	missense	c.1891A>G	p.Arg631Gly
2:25243944:CT:C	NA	frameshift	c.1889delA	p.Lys630fs
2:25243944:CTT:C	NA	frameshift	c.1888_1889delAA	p.Lys630fs
2:25243945:T:TTC	NA	frameshift	c.1887_1888dupG A	p.Lys630fs
2:25243946:T:A	NA	stop_gained	c.1888A>T	p.Lys630*
2:25243947:CT:C	NA	frameshift	c.1886delA	p.Glu629fs
2:25243947:CTCA GCTGGGACAGGT GGGTAAA:C	NA	frameshift	c.1865_1886delTTT ACCCACCTGTCCCA GCTGA	p.Val622fs
2:25243949:C:A	NA	stop_gained	c.1885G>T	p.Glu629*
2:25243949:C:CA	NA	frameshift	c.1884dupT	p.Glu629fs
2:25243949:CAG: C	NA	frameshift	c.1883_1884delCT	p.Ala628fs
2:25243951:G:T	rs149876809	missense	c.1883C>A	p.Ala628Asp
2:25243951:GC:G	NA	frameshift	c.1882delG	p.Ala628fs
2:25243953:T:TG	NA	frameshift	c.1880dupC	p.Ala628fs
2:25243953:TG:T	NA	frameshift	c.1880delC	p.Pro627fs
2:25243954:GGG ACAGGTGGGTAA ACCTTT:G	NA	frameshift	c.1860_1879delAA AGGTTTACCCACCT GTCC	p.Lys621fs
2:25243956:GA:G	NA	frameshift	c.1877delT	p.Val626fs
2:25243956:GAC AGGTGGGTAA:G	NA	inframe_indel	c.1866_1877delTT ACCCACCTGT	p.Tyr623_Val626 del
2:25243957:A:C	NA	missense	c.1877T>G	p.Val626Gly
2:25243957:A:T	NA	missense	c.1877T>A	p.Val626Asp

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2:25243957:AC:A	NA	frameshift	c.1876delG	p.Val626fs
2:25243958:CA:C	NA	frameshift	c.1875delT	p.Val626fs
2:25243958:CAG GTGGGTAA:C	NA	frameshift	c.1866_1875delTT ACCCACCT	p.Tyr623fs
2:25243959:A:AG GTGG	NA	frameshift	c.1870_1874dupCC ACC	p.Val626fs
2:25243959:AG:A	NA	frameshift	c.1874delC	p.Pro625fs
2:25243960:G:C	NA	missense	c.1874C>G	p.Pro625Arg
2:25243960:G:GG T	NA	frameshift	c.1872_1873dupAC	p.Pro625fs
2:25243961:G:GT	NA	frameshift	c.1872dupA	p.Pro625fs
2:25243961:GT:G	rs745381125	frameshift	c.1872delA	p.Pro625fs
2:25243962:TG:T	NA	frameshift	c.1871delC	p.Pro624fs
2:25243963:GGG TAA:G	NA	frameshift	c.1866_1870delTT ACC	p.Tyr623fs
2:25243963:GGG TAAA:G	rs1394353623	inframe_indel	c.1865_1870delTTT ACC	p.Val622_Pro624 delinsAla
2:25243964:G:GG TAAACCTTTGGAG GGTCCTAAGCAT	NA	frameshift	c.1869_1870insAT GCTTAGGACCCTCC AAAGGTTTAC	p.Pro624fs
2:25243964:G:GG TTTA	NA	frameshift	c.1869_1870insTA AAC	p.Pro624fs
2:25243965:G:C	NA	stop_gained	c.1869C>G	p.Tyr623*
2:25243965:G:GT T	NA	frameshift	c.1868_1869insAA	p.Tyr623fs
2:25243965:G:T	NA	stop_gained	c.1869C>A	p.Tyr623*
2:25243965:GT:G	NA	frameshift	c.1868delA	p.Tyr623fs
2:25243966:T:TA	NA	frameshift	c.1867dupT	p.Tyr623fs
2:25243966:T:TA ACCC	NA	frameshift	c.1867_1868insGG GTT	p.Tyr623fs
2:25243966:TA:T	NA	frameshift	c.1867delT	p.Tyr623fs
2:25243966:TAAA CC:T	NA	frameshift	c.1863_1867delGG TTT	p.Val622fs

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2:25243968:A:AA C	rs1477019156	frameshift	c.1864_1865dupGT	p.Tyr623fs
2:25243968:A:AT TAC	NA	frameshift	c.1865_1866insGT AA	p.Tyr623fs
2:25243969:AC:A	NA	frameshift	c.1864delG	p.Val622fs
2:25243969:ACCT T:A	NA	frameshift	c.1861_1864delAA GG	p.Lys621fs
2:25243971:CT:C	NA	frameshift	c.1862delA	p.Lys621fs
2:25243973:T:A	NA	stop_gained	c.1861A>T	p.Lys621*
2:25243974:TG:T	NA	frameshift	c.1859delC	p.Pro620fs
2:25243976:GA:G	NA	frameshift	c.1857delT	p.Pro620fs
2:25243977:AG:A	NA	frameshift	c.1856delC	p.Pro619fs
2:25243980:GT:G	NA	frameshift	c.1853delA	p.Asp618fs
2:25243981:TC:T	NA	frameshift	c.1852delG	p.Asp618fs
2:25243983:C:T	rs866791048	splice_acceptor	c.1852-1G>A	
2:25244151:TCA: T	rs1228601968	splice_donor	c.1851+2_1851+3d elTG	
2:25244152:CA:C	NA	splice_donor	c.1851+2delT	
2:25244152:CACA AATT:C	NA	frameshift	c.1847_1851+2del AATTTGT	p.Glu616fs
2:25244153:A:C	NA	splice_donor	c.1851+2T>G	
2:25244153:A:G	rs757785566	splice_donor	c.1851+2T>C	
2:25244153:A:T	rs757785566	splice_donor	c.1851+2T>A	
2:25244153:AC:A	NA	splice_donor	c.1851+1delG	
2:25244153:ACAA AT:A	NA	frameshift	c.1848_1851+1del ATTTG	p.Phe617fs
2:25244154:C:A	NA	splice_donor	c.1851+1G>T	
2:25244154:C:G	NA	splice_donor	c.1851+1G>C	
2:25244154:C:T	rs928051436	splice_donor	c.1851+1G>A	
2:25244154:CA:C	NA	frameshift	c.1851delT	p.Phe617fs
2:25244157:AT:A	NA	frameshift	c.1848delA	p.Glu616fs
2:25244158:T:TG	NA	frameshift	c.1847_1848insC	p.Glu616fs

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2:25244159:T:A	NA	missense	c.1847A>T	p.Glu616Val
2:25244159:TC:T	rs1409292254	frameshift	c.1846delG	p.Glu616fs
2:25244160:C:A	rs779600073	stop_gained	c.1846G>T	p.Glu616*
2:25244161:CT:C	NA	frameshift	c.1844delA	p.Gln615fs
2:25244162:TG:T	NA	frameshift	c.1843delC	p.Gln615fs
2:25244162:TGGT C:T	NA	frameshift	c.1840_1843delGA CC	p.Asp614fs
2:25244163:G:A	rs768548187	stop_gained	c.1843C>T	p.Gln615*
2:25244163:G:GG TCGT	NA	frameshift	c.1838_1842dupAC GAC	p.Gln615fs
2:25244164:GT:G	NA	frameshift	c.1841delA	p.Asp614fs
2:25244165:TC:T	rs757911864	frameshift	c.1840delG	p.Asp614fs
2:25244166:CG:C	NA	frameshift	c.1839delC	p.His613fs
2:25244167:GT:G	rs1431450984	frameshift	c.1838delA	p.His613fs
2:25244167:GTG GT:G	NA	frameshift	c.1835_1838delAC CA	p.Asn612fs
2:25244167:GTG GTTATTAGCGAAG AACATC:G	NA	inframe_indel	c.1818_1838delGA TGTTCTTCGCTAAT AACCA	p.Gln606_Asn612 del
2:25244168:T:TG	NA	frameshift	c.1837dupC	p.His613fs
2:25244168:TG:T	NA	frameshift	c.1837delC	p.His613fs
2:25244168:TGGT TATTAGC:T	NA	frameshift	c.1828_1837delGC TAATAACC	p.Ala610fs
2:25244170:G:GT T	NA	frameshift	c.1834_1835dupAA	p.Asn612fs
2:25244170:GT:G	NA	frameshift	c.1835delA	p.Asn612fs
2:25244170:GTT: G	NA	frameshift	c.1834_1835delAA	p.Asn612fs
2:25244170:GTTA T:G	NA	frameshift	c.1832_1835delAT AA	p.Asn611fs
2:25244172:TA:T	NA	frameshift	c.1833delT	p.Asn611fs
2:25244172:TATT AGCG:T	NA	frameshift	c.1827_1833delCG CTAAT	p.Phe609fs

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2:25244172:TATT AGCGA:T	NA	frameshift	c.1826_1833delTC GCTAAT	p.Phe609fs
2:25244173:AT:A	NA	frameshift	c.1832delA	p.Asn611fs
2:25244174:TTAG CG:T	NA	frameshift	c.1827_1831delCG CTA	p.Phe609fs
2:25244176:AG:A	NA	frameshift	c.1829delC	p.Ala610fs
2:25244177:G:C	rs930025818	missense	c.1829C>G	p.Ala610Gly
2:25244178:C:CG	NA	frameshift	c.1827dupC	p.Ala610fs
2:25244178:C:T	NA	missense	c.1828G>A	p.Ala610Thr
2:25244178:CG:C		frameshift	c.1827delC	p.Phe609fs
2:25244179:GA:G	NA	frameshift	c.1826delT	p.Phe609fs
2:25244181:AG:A	NA	frameshift	c.1824delC	p.Phe609fs
2:25244182:G:GC	NA	frameshift	c.1823_1824insG	p.Phe608fs
2:25244182:G:GT	NA	frameshift	c.1823_1824insA	p.Phe608fs
2:25244182:GA:G	NA	frameshift	c.1823delT	p.Phe608fs
2:25244183:AAC: A	NA	frameshift	c.1821_1822delGT	p.Met607fs
2:25244184:A:AC ATCT	NA	frameshift	c.1817_1821dupA GATG	p.Phe608fs
2:25244184:A:AC ATCTGGAGC	NA	frameshift	c.1812_1821dupGC TCCAGATG	p.Phe608fs
2:25244185:CATC TG:C	NA	frameshift	c.1816_1820delCA GAT	p.Gln606fs
2:25244188:CT:C	NA	frameshift	c.1817delA	p.Gln606fs
2:25244188:CTG: C	NA	frameshift	c.1816_1817delCA	p.Gln606fs
2:25244189:T:TG GAGCCG	NA	frameshift	c.1810_1816dupCG GCTCC	p.Gln606fs
2:25244189:TG:T	NA	frameshift	c.1816delC	p.Gln606fs
2:25244190:G:A	rs1396358001	stop_gained	c.1816C>T	p.Gln606*
2:25244193:GC:G	NA	frameshift	c.1812delG	p.Leu605fs
2:25244195:C:G	NA	missense	c.1811G>C	p.Arg604Pro
2:25244195:C:T	rs759818409	missense	c.1811G>A	p.Arg604Gln
2:25244195:CG:C	rs779782698	frameshift	c.1810delC	p.Arg604fs

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2:25244196:G:A	rs1368287759	missense	c.1810C>T	p.Arg604Trp
2:25244196:G:C	NA	missense	c.1810C>G	p.Arg604Gly
2:25244198:GA:G	NA	frameshift	c.1807delT	p.Ser603fs
2:25244199:AG:A	NA	frameshift	c.1806delC	p.Ser603fs
2:25244199:AGG: A	NA	frameshift	c.1805_1806delCC	p.Pro602fs
2:25244199:AGG GC:A	NA	frameshift	c.1803_1806delGC CC	p.Trp601fs
2:25244202:G:T	NA	missense	c.1804C>A	p.Pro602Thr
2:25244202:GC:G	NA	frameshift	c.1803delG	p.Trp601fs
2:25244202:GCCA GT:G	NA	frameshift	c.1799_1803delAC TGG	p.Asp600fs
2:25244203:C:T	rs767692203	stop_gained	c.1803G>A	p.Trp601*
2:25244203:CCA: C	NA	frameshift	c.1801_1802delTG	p.Trp601fs
2:25244204:C:T	rs941325374	stop_gained	c.1802G>A	p.Trp601*
2:25244204:CA:C	NA	frameshift	c.1801delT	p.Trp601fs
2:25244205:A:AG	NA	frameshift	c.1800dupC	p.Trp601fs
2:25244205:A:G	NA	missense	c.1801T>C	p.Trp601Arg
2:25244205:AG:A	NA	frameshift	c.1800delC	p.Trp601fs
2:25244206:G:GT CCTC	NA	frameshift	c.1795_1799dupG AGGA	p.Asp600fs
2:25244208:C:CC T	NA	frameshift	c.1796_1797dupA G	p.Asp600fs
2:25244209:CT:C	NA	frameshift	c.1796delA	p.Glu599fs
2:25244211:C:A	NA	stop_gained	c.1795G>T	p.Glu599*
2:25244211:C:CT	NA	frameshift	c.1794dupA	p.Glu599fs
2:25244212:T:TC	NA	frameshift	c.1793dupG	p.Glu599fs
2:25244212:TCGC :T	NA	inframe_indel	c.1791_1793delGC G	p.Arg598del
2:25244213:C:T	rs764679587	missense	c.1793G>A	p.Arg598Gln
2:25244214:G:A	rs568207978	stop_gained	c.1792C>T	p.Arg598*
2:25244214:G:C	NA	missense	c.1792C>G	p.Arg598Gly

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2:25244216:CG:C	NA	frameshift	c.1789delC	p.Arg597fs
2:25244216:CGCC GCAGCA:C	NA	inframe_indel	c.1781_1789delTG CTGCGGC	p.Leu594_Arg596 del
2:25244217:G:A	rs757492795	missense	c.1789C>T	p.Arg597Trp
2:25244217:G:GC	NA	frameshift	c.1788dupG	p.Arg597fs
2:25244217:GC:G	NA	frameshift	c.1788delG	p.Arg597fs
2:25244218:C:CC GCAG	NA	frameshift	c.1783_1787dupCT GCG	p.Arg597fs
2:25244218:C:CC GCAGCAGCCCGTA GT	NA	frameshift	c.1787_1788insACT ACGGGCTGCTGCG	p.Arg597fs
2:25244219:C:T	rs899717364	missense	c.1787G>A	p.Arg596Gln
2:25244219:CGCA GCAGCCCGTAG:C	NA	frameshift	c.1773_1786delCT ACGGGCTGCTGC	p.Tyr592fs
2:25244220:G:GC	NA	frameshift	c.1785dupG	p.Arg596fs
2:25244220:GC:G	NA	frameshift	c.1785delG	p.Arg596fs
2:25244221:CA:C	NA	frameshift	c.1784delT	p.Leu595fs
2:25244222:A:C	NA	missense	c.1784T>G	p.Leu595Arg
2:25244222:A:G	rs1483256801	missense	c.1784T>C	p.Leu595Pro
2:25244223:G:GC AGC	NA	frameshift	c.1779_1782dupGC TG	p.Leu595fs
2:25244223:GC:G	NA	frameshift	c.1782delG	p.Leu595fs
2:25244224:CA:C	NA	frameshift	c.1781delT	p.Leu594fs
2:25244225:AGCC CGTAGGTACCTT GTGCCC:A	NA	frameshift	c.1759_1780delGG GCACAAGGGTACCT ACGGGC	p.Gly587fs
2:25244226:GC:G	NA	frameshift	c.1779delG	p.Leu594fs
2:25244229:C:CG TAG	NA	frameshift	c.1773_1776dupCT AC	p.Gly593fs
2:25244229:C:T	rs755573228	missense	c.1777G>A	p.Gly593Arg
2:25244229:CG:C	NA	frameshift	c.1776delC	p.Tyr592fs
2:25244230:G:C	rs758063797	stop_gained	c.1776C>G	p.Tyr592*
2:25244230:G:GT C	NA	frameshift	c.1775_1776insGA	p.Tyr592fs

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2:25244230:G:T	rs758063797	stop_gained	c.1776C>A	p.Tyr592*
2:25244231:TA:T	NA	frameshift	c.1774delT	p.Tyr592fs
2:25244231:TAG GTACCCTTGTGCC CGCAC:T	NA	frameshift	c.1755_1774delGT GCGGGCACAAGGG TACCT	p.Met585fs
2:25244232:A:AG	NA	frameshift	c.1773dupC	p.Tyr592fs
2:25244234:G:A	NA	missense	c.1772C>T	p.Thr591Ile
2:25244235:TACC CTTG:T	NA	frameshift	c.1764_1770delCA AGGGT	p.His588fs
2:25244235:TACC CTTGTGCCCGCAC ATGTAGC:T	NA	frameshift	c.1748_1770delGC TACATGTGCGGGCA CAAGGGT	p.Cys583fs
2:25244236:AC:A	NA	frameshift	c.1769delG	p.Gly590fs
2:25244236:ACCC TTGT:A	NA	frameshift	c.1763_1769delAC AAGGG	p.His588fs
2:25244239:CT:C	NA	frameshift	c.1766delA	p.Lys589fs
2:25244240:T:C	rs1324043719	missense	c.1766A>G	p.Lys589Arg
2:25244240:TTG: T	NA	frameshift	c.1764_1765delCA	p.His588fs
2:25244240:TTGT GC:T	NA	frameshift	c.1761_1765delGC ACA	p.His588fs
2:25244242:GT:G	NA	frameshift	c.1763delA	p.His588fs
2:25244242:GTGC CCGCACAT:G	NA	frameshift	c.1753_1763delAT GTGCGGGCA	p.Met585fs
2:25244243:T:TG CCC	NA	frameshift	c.1759_1762dupG GGC	p.His588fs
2:25244243:TG:T	NA	frameshift	c.1762delC	p.His588fs
2:25244244:GC:G	rs1313980878	frameshift	c.1761delG	p.His588fs
2:25244246:C:A	NA	missense	c.1760G>T	p.Gly587Val
2:25244246:C:G	NA	missense	c.1760G>C	p.Gly587Ala
2:25244247:CGCA CATG:C	NA	frameshift	c.1752_1758delCA TGTGC	p.Tyr584fs
2:25244248:G:C	NA	missense	c.1758C>G	p.Cys586Trp

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2:25244248:G:GC A	NA	frameshift	c.1756_1757dupTG	p.Gly587fs
2:25244248:G:T	rs746498722	stop_gained	c.1758C>A	p.Cys586*
2:25244248:GCA: G	NA	frameshift	c.1756_1757delTG	p.Cys586fs
2:25244249:C:CA	NA	frameshift	c.1756dupT	p.Cys586fs
2:25244249:C:G	NA	missense	c.1757G>C	p.Cys586Ser
2:25244249:C:T	rs754506713	missense	c.1757G>A	p.Cys586Tyr
2:25244250:A:AC	rs1371954044	frameshift	c.1755dupG	p.Cys586fs
2:25244250:A:C	NA	missense	c.1756T>G	p.Cys586Gly
2:25244250:A:G	NA	missense	c.1756T>C	p.Cys586Arg
2:25244250:AC:A	rs1376463207	frameshift	c.1755delG	p.Met585fs
2:25244250:ACAT GTAGCAGTTC:A	NA	frameshift	c.1743_1755delGA ACTGCTACATG	p.Trp581fs
2:25244251:CA:C	NA	frameshift	c.1754delT	p.Met585fs
2:25244251:CATG TAGCA:C	NA	frameshift	c.1747_1754delTG CTACAT	p.Cys583fs
2:25244251:CATG TAGCAGT:C	NA	frameshift	c.1745_1754delAC TGCTACAT	p.Asn582fs
2:25244252:A:AT G	NA	frameshift	c.1752_1753dupCA	p.Met585fs
2:25244252:AT:A	NA	frameshift	c.1753delA	p.Met585fs
2:25244252:ATGT AGCAG:A	NA	frameshift	c.1746_1753delICT GCTACA	p.Cys583fs
2:25244253:T:C	NA	missense	c.1753A>G	p.Met585Val
2:25244253:TG:T	NA	frameshift	c.1752delC	p.Tyr584fs
2:25244253:TGTA GCAGTTCAG:T	NA	frameshift	c.1740_1752delICT GGAAGTCTAC	p.Trp581fs
2:25244253:TGTA GCAGTTCAGG:T	NA	frameshift	c.1739_1752delICC TGGAAGTCTAC	p.Pro580fs
2:25244254:G:C	NA	stop_gained	c.1752C>G	p.Tyr584*
2:25244254:G:GT	NA	frameshift	c.1751dupA	p.Tyr584fs
2:25244254:G:T	NA	stop_gained	c.1752C>A	p.Tyr584*
2:25244254:GT:G	NA	frameshift	c.1751delA	p.Tyr584fs

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2:25244255:T:C	NA	missense	c.1751A>G	p.Tyr584Cys
2:25244256:A:G	NA	missense	c.1750T>C	p.Tyr584His
2:25244257:G:C	NA	missense	c.1749C>G	p.Cys583Trp
2:25244257:G:T	NA	stop_gained	c.1749C>A	p.Cys583*
2:25244257:GC:G	NA	frameshift	c.1748delG	p.Cys583fs
2:25244258:C:G	NA	missense	c.1748G>C	p.Cys583Ser
2:25244258:C:T	rs748082838	missense	c.1748G>A	p.Cys583Tyr
2:25244259:A:G	NA	missense	c.1747T>C	p.Cys583Arg
2:25244259:A:T	NA	missense	c.1747T>A	p.Cys583Ser
2:25244259:AG:A	NA	frameshift	c.1746delC	p.Cys583fs
2:25244260:G:GT	NA	frameshift	c.1745dupA	p.Asn582fs
2:25244260:GT:G	NA	frameshift	c.1745delA	p.Asn582fs
2:25244260:GTTC CAGGGGTC:G	NA	frameshift	c.1735_1745delGA CCCCTGGAA	p.Asp579fs
2:25244262:TC:T	NA	frameshift	c.1743delG	p.Trp581fs
2:25244263:C:A	rs769419803	missense	c.1743G>T	p.Trp581Cys
2:25244263:C:CC A	NA	frameshift	c.1741_1742dupTG	p.Trp581fs
2:25244263:C:G	rs769419803	missense	c.1743G>C	p.Trp581Cys
2:25244263:C:T	rs769419803	stop_gained	c.1743G>A	p.Trp581*
2:25244264:C:A	NA	missense	c.1742G>T	p.Trp581Leu
2:25244264:C:G	NA	missense	c.1742G>C	p.Trp581Ser
2:25244264:C:T	rs1050697275	stop_gained	c.1742G>A	p.Trp581*
2:25244264:CA:C	NA	frameshift	c.1741delT	p.Trp581fs
2:25244264:CAG GG:C	NA	frameshift	c.1738_1741delCC CT	p.Pro580fs
2:25244264:CAG GGGTCTTCCTTAA TGGCTG:C	NA	inframe_indel	c.1721_1741delCA GCCATTAAGGAAG ACCCCT	p.Ala574_Trp581 delinsGly
2:25244265:A:AG	NA	frameshift	c.1740dupC	p.Trp581fs
2:25244265:A:AG G	NA	frameshift	c.1739_1740dupCC	p.Trp581fs
2:25244265:A:C	rs1342803588	missense	c.1741T>G	p.Trp581Gly

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2:25244265:A:G	NA	missense	c.1741T>C	p.Trp581Arg
2:25244265:A:T	NA	missense	c.1741T>A	p.Trp581Arg
2:25244265:AG:A	NA	frameshift	c.1740delC	p.Trp581fs
2:25244266:GGG GTCTTCCT:G	NA	frameshift	c.1730_1739delAG GAAGACCC	p.Lys577fs
2:25244266:GGG GTCTTCCTTAATG GCTGCCT:G	NA	frameshift	c.1718_1739delAG GCAGCCATTAAGGA AGACCC	p.Gln573fs
2:25244267:G:A	rs773048923	missense	c.1739C>T	p.Pro580Leu
2:25244267:G:T	NA	missense	c.1739C>A	p.Pro580His
2:25244267:GGG TC:G	NA	frameshift	c.1735_1738delGA CC	p.Asp579fs
2:25244268:G:A	NA	missense	c.1738C>T	p.Pro580Ser
2:25244269:GT:G	NA	frameshift	c.1736delA	p.Asp579fs
2:25244269:GTCT TC:G	NA	frameshift	c.1732_1736delGA AGA	p.Glu578fs
2:25244270:T:A	NA	missense	c.1736A>T	p.Asp579Val
2:25244270:T:C	NA	missense	c.1736A>G	p.Asp579Gly
2:25244270:TCTT C:T	NA	frameshift	c.1732_1735delGA AG	p.Glu578fs
2:25244270:TCTT CCTTAATGGCTGC: T	NA	frameshift	c.1720_1735delGC AGCCATTAAGGAA G	p.Ala574fs
2:25244271:C:A	NA	missense	c.1735G>T	p.Asp579Tyr
2:25244271:C:CT	NA	frameshift	c.1734dupA	p.Asp579fs
2:25244271:C:T	NA	missense	c.1735G>A	p.Asp579Asn
2:25244271:CT:C	NA	frameshift	c.1734delA	p.Asp579fs
2:25244272:T:TA GAC	NA	frameshift	c.1733_1734insGT CT	p.Asp579fs
2:25244272:TTC:T	NA	frameshift	c.1732_1733delGA	p.Glu578fs
2:25244272:TTCC: T	NA	inframe_indel	c.1731_1733delGG A	p.Glu578del
2:25244273:T:C	NA	missense	c.1733A>G	p.Glu578Gly
2:25244273:T:TC	NA	frameshift	c.1732dupG	p.Glu578fs

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2:25244273:TC:T	NA	frameshift	c.1732delG	p.Glu578fs
2:25244273:TCC:T	NA	frameshift	c.1731_1732delGG	p.Glu578fs
2:25244273:TCCT TAATGGCTG:T	NA	inframe_indel	c.1721_1732delCA GCCATTAAGG	p.Ala574_Lys577del
2:25244274:C:A	NA	stop_gained	c.1732G>T	p.Glu578*
2:25244275:C:CT	NA	frameshift	c.1730dupA	p.Glu578fs
2:25244275:CTTA A:C	NA	frameshift	c.1727_1730delTT AA	p.Ile576fs
2:25244277:T:TA	NA	frameshift	c.1728dupT	p.Lys577fs
2:25244277:TA:T	NA	frameshift	c.1728delT	p.Lys577fs
2:25244278:AATG GCTGCC:A	NA	inframe_indel	c.1719_1727delGG CAGCCAT	p.Gln573_Ile576delinsHis
2:25244278:AATG GCTGCCTGGGCA GCCCCCG:A	NA	frameshift	c.1706_1727delCG GGGGCTGCCCAGG CAGCCAT	p.Pro569fs
2:25244279:A:C	rs1247716811	missense	c.1727T>G	p.Ile576Ser
2:25244279:A:G	NA	missense	c.1727T>C	p.Ile576Thr
2:25244279:A:T	NA	missense	c.1727T>A	p.Ile576Asn
2:25244279:AT:A	NA	frameshift	c.1726delA	p.Ile576fs
2:25244279:ATG GCTGCC:A	NA	frameshift	c.1719_1726delGG CAGCCA	p.Gln573fs
2:25244279:ATG GCTGCCTGGGCA GC:A	NA	frameshift	c.1711_1726delGC TGCCCAGGCAGCCA	p.Ala571fs
2:25244280:T:C	NA	missense	c.1726A>G	p.Ile576Val
2:25244280:T:G	NA	missense	c.1726A>C	p.Ile576Leu
2:25244280:T:TA A	NA	frameshift	c.1725_1726insTT	p.Ile576fs
2:25244280:TG:T	NA	frameshift	c.1725delC	p.Ile576fs
2:25244280:TGGC TGCCTGGGCAGCC C:T	NA	frameshift	c.1709_1725delGG GCTGCCCAGGCAGC C	p.Gly570fs

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2:25244280:TGGC TGCCTGGGCAGCC CCC:T	rs770302280	frameshift	c.1707_1725delGG GGGCTGCCCAGGC AGCC	p.Gly570fs
2:25244281:GGCT GCC:G	NA	inframe_indel	c.1719_1724delGG CAGC	p.Gln573_Ala575 delinsHis
2:25244281:GGCT GCCTGGGCA:G	NA	inframe_indel	c.1713_1724delTG CCCAGGCAGC	p.Ala572_Ala575 del
2:25244281:GGCT GCCTGGGCAGCC CCC:G	NA	inframe_indel	c.1707_1724delGG GGGCTGCCCAGGC AGC	p.Gly570_Ala575 del
2:25244282:G:A	NA	missense	c.1724C>T	p.Ala575Val
2:25244282:GCTG CCTGGGCAGC:G	NA	frameshift	c.1711_1723delGC TGCCCAGGCAG	p.Ala571fs
2:25244282:GCTG CCTGGGCAGCC:G	NA	frameshift	c.1710_1723delGG CTGCCCAGGCAG	p.Ala571fs
2:25244282:GCTG CCTGGGCAGCCC: G	NA	inframe_indel	c.1709_1723delGG GCTGCCCAGGCAG	p.Gly570_Ala574 del
2:25244282:GCTG CCTGGGCAGCCCC CGGCCCCACC:G	NA	frameshift	c.1698_1723delGG TGGGGCCGGGGGC TGCCCAGGCAG	p.Leu566fs
2:25244283:C:G	NA	missense	c.1723G>C	p.Ala575Pro
2:25244283:CT:C	NA	frameshift	c.1722delA	p.Ala575fs
2:25244284:TG:T	NA	frameshift	c.1721delC	p.Ala574fs
2:25244284:TGCC TGGGCA:T	rs773643031	inframe_indel	c.1713_1721delTG CCCAGGC	p.Ala572_Ala574 del
2:25244284:TGCC TGGGCAGCC:T	NA	inframe_indel	c.1710_1721delGG CTGCCCAGGC	p.Ala571_Ala574 del
2:25244284:TGCC TGGGCAGCCCC: T	NA	inframe_indel	c.1707_1721delGG GGGCTGCCCAGGC	p.Gly570_Ala574 del
2:25244285:GC:G	NA	frameshift	c.1720delG	p.Ala574fs
2:25244285:GCCT GGGCAGC:G	NA	frameshift	c.1711_1720delGC TGCCCAGG	p.Ala571fs

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2:25244285:GCCT GGGCAGCC:G	NA	frameshift	c.1710_1720delGG CTGCCCAGG	p.Ala571fs
2:25244286:C:T	NA	missense	c.1720G>A	p.Ala574Thr
2:25244286:CCTG GGCAG:C	NA	frameshift	c.1712_1719delICT GCCCAG	p.Ala571fs
2:25244286:CCTG GGCAGCCCCG:C	NA	frameshift	c.1706_1719delICG GGGGCTGCCCAG	p.Pro569fs
2:25244287:C:A		missense	c.1719G>T	p.Gln573His
2:25244287:C:CC A	NA	frameshift	c.1718_1719insTG	p.Gln573fs
2:25244287:CTGG :C	NA	inframe_indel	c.1716_1718delICC A	p.Gln573del
2:25244287:CTGG GCAG:C	rs763053459	frameshift	c.1712_1718delICT GCCCCA	p.Ala571fs
2:25244288:T:C	NA	missense	c.1718A>G	p.Gln573Arg
2:25244288:TG:T	NA	frameshift	c.1717delIC	p.Gln573fs
2:25244288:TGG GCAGCCCC:T	NA	frameshift	c.1708_1717delGG GGCTGCCC	p.Gly570fs
2:25244289:G:A	rs902965419	stop_gained	c.1717C>T	p.Gln573*
2:25244289:G:GG GCAGCCCCA	NA	frameshift	c.1716_1717insTG GGGCTGCC	p.Gln573fs
2:25244289:GGG CAGCC:G	NA	frameshift	c.1710_1716delGG CTGCC	p.Ala571fs
2:25244289:GGG CAGCCCCC:G	NA	frameshift	c.1707_1716delGG GGGCTGCC	p.Gly570fs
2:25244290:G:GG CA	NA	inframe_indel	c.1713_1715dupTG C	p.Ala572dup
2:25244290:GGC: G	NA	frameshift	c.1714_1715delGC	p.Ala572fs
2:25244290:GGC A:G	rs1351573938	inframe_indel	c.1713_1715delITG C	p.Ala572del
2:25244290:GGC AGC:G	NA	frameshift	c.1711_1715delGC TGC	p.Ala571fs

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2:25244290:GGC AGCCCC:G	NA	frameshift	c.1708_1715delGG GGCTGC	p.Gly570fs
2:25244291:G:T	NA	missense	c.1715C>A	p.Ala572Asp
2:25244291:GCA GCCC:G	NA	inframe_indel	c.1709_1714delGG GCTG	p.Gly570_Ala571 del
2:25244292:C:T	NA	missense	c.1714G>A	p.Ala572Thr
2:25244292:CA:C	NA	frameshift	c.1713delT	p.Ala572fs
2:25244293:AG:A	NA	frameshift	c.1712delC	p.Ala571fs
2:25244294:G:GC		frameshift	c.1711dupG	p.Ala571fs
2:25244294:G:T	NA	missense	c.1712C>A	p.Ala571Asp
2:25244294:GC:G		frameshift	c.1711delG	p.Ala571fs
2:25244295:C:G	NA	missense	c.1711G>C	p.Ala571Pro
2:25244295:C:T	NA	missense	c.1711G>A	p.Ala571Thr
2:25244297:C:A	NA	missense	c.1709G>T	p.Gly570Val
2:25244297:C:G	NA	missense	c.1709G>C	p.Gly570Ala
2:25244297:C:T	NA	missense	c.1709G>A	p.Gly570Glu
2:25244298:C:T	NA	missense	c.1708G>A	p.Gly570Arg
2:25244300:G:A	rs772310511	missense	c.1706C>T	p.Pro569Leu
2:25244300:G:C	NA	missense	c.1706C>G	p.Pro569Arg
2:25244301:G:GC A	NA	frameshift	c.1704_1705insTG	p.Pro569fs
2:25244301:GC:G	NA	frameshift	c.1704delG	p.Pro569fs
2:25244301:GCC: G	NA	frameshift	c.1703_1704delGG	p.Gly568fs
2:25244301:GCCC CACCAAGAGGTCC ACACACT:G	NA	frameshift	c.1682_1704delAG TGTGTGGACCTCTT GGTGGGG	p.Glu561fs
2:25244302:CCCC A:C	NA	frameshift	c.1700_1703delTG GG	p.Val567fs
2:25244303:C:T	NA	missense	c.1703G>A	p.Gly568Glu
2:25244303:CCCA :C	rs1361235617	inframe_indel	c.1700_1702delTG G	p.Val567del
2:25244304:CCAC CAAGAGGTCCA:C	NA	frameshift	c.1688_1701delTG GACCTCTTGGTG	p.Val563fs

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2:25244305:CA:C	NA	frameshift	c.1700delT	p.Val567fs
2:25244306:A:AC	NA	frameshift	c.1699dupG	p.Val567fs
2:25244306:AC:A	NA	frameshift	c.1699delG	p.Val567fs
2:25244307:CCAA G:C	NA	frameshift	c.1695_1698delCTT G	p.Leu566fs
2:25244308:C:CA AGA	NA	frameshift	c.1694_1697dupTC TT	p.Leu566fs
2:25244308:C:CC A	NA	frameshift	c.1697_1698insTG	p.Leu566fs
2:25244308:CA:C	NA	frameshift	c.1697delT	p.Leu566fs
2:25244309:A:T	NA	stop_gained	c.1697T>A	p.Leu566*
2:25244311:GA:G	NA	frameshift	c.1694delT	p.Leu565fs
2:25244312:A:G	rs1040053651	missense	c.1694T>C	p.Leu565Pro
2:25244312:AG:A	rs1303060010	frameshift	c.1693delC	p.Leu565fs
2:25244312:AGG TC:A	NA	frameshift	c.1690_1693delGA CC	p.Asp564fs
2:25244314:G:GT CCAC	NA	frameshift	c.1687_1691dupGT GGA	p.Asp564fs
2:25244314:GT:G	NA	frameshift	c.1691delA	p.Asp564fs
2:25244315:T:TC	NA	frameshift	c.1690dupG	p.Asp564fs
2:25244315:TC:T	NA	frameshift	c.1690delG	p.Asp564fs
2:25244315:TCC:T	NA	frameshift	c.1689_1690delGG	p.Asp564fs
2:25244315:TCCA CACACTCCA:T	NA	inframe_indel	c.1679_1690delITG GAGTGTGTGG	p.Val560_Val563d el
2:25244315:TCCA CACACTCCACGCA AAAGCA:T	NA	frameshift	c.1669_1690delITG CTTTTTCGTGGAGT GTGTGG	p.Cys557fs
2:25244316:C:CC A	NA	frameshift	c.1688_1689dupTG	p.Asp564fs
2:25244316:C:CC ACA	NA	frameshift	c.1686_1689dupTG TG	p.Asp564fs
2:25244316:CCA: C	NA	frameshift	c.1688_1689delITG	p.Val563fs
2:25244317:C:CA	NA	frameshift	c.1688dupT	p.Asp564fs

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2:25244318:A:AC ACACTC	NA	frameshift	c.1681_1687dupG AGTGTG	p.Val563fs
2:25244318:A:T	NA	missense	c.1688T>A	p.Val563Glu
2:25244318:AC:A	NA	frameshift	c.1687delG	p.Val563fs
2:25244318:ACAC ACTCCACG:A	NA	frameshift	c.1677_1687delCG TGGAGTGTG	p.Val560fs
2:25244319:C:T	rs1299203502	missense	c.1687G>A	p.Val563Met
2:25244319:CA:C	NA	frameshift	c.1686delT	p.Cys562fs
2:25244320:ACAC TCCAC:A	NA	frameshift	c.1678_1685delGT GGAGTG	p.Val560fs
2:25244321:C:A	rs1218552501	missense	c.1685G>T	p.Cys562Phe
2:25244321:C:G	NA	missense	c.1685G>C	p.Cys562Ser
2:25244321:C:T	rs1218552501	missense	c.1685G>A	p.Cys562Tyr
2:25244321:CA:C	NA	frameshift	c.1684delT	p.Cys562fs
2:25244322:A:G	rs1057520788	missense	c.1684T>C	p.Cys562Arg
2:25244322:AC:A	NA	frameshift	c.1683delG	p.Glu561fs
2:25244322:ACTC CACGCAAAAG:A	NA	frameshift	c.1671_1683delCTT TTGCGTGGAG	p.Phe558fs
2:25244324:TC:T	NA	frameshift	c.1681delG	p.Glu561fs
2:25244324:TCCA CG:T	NA	frameshift	c.1677_1681delCG TGG	p.Cys559fs
2:25244324:TCCA CGCAAAAG:T	NA	frameshift	c.1671_1681delCTT TTGCGTGG	p.Cys557fs
2:25244325:C:A	NA	stop_gained	c.1681G>T	p.Glu561*
2:25244325:CCA: C	NA	frameshift	c.1679_1680delTG	p.Val560fs
2:25244325:CCAC G:C	NA	frameshift	c.1677_1680delCG TG	p.Cys559fs
2:25244326:CA:C	NA	frameshift	c.1679delT	p.Val560fs
2:25244327:AC:A	NA	frameshift	c.1678delG	p.Val560fs
2:25244327:ACGC AAAAG:A	NA	frameshift	c.1671_1678delCTT TTGCG	p.Phe558fs
2:25244328:CG:C	NA	frameshift	c.1677delC	p.Cys559fs
2:25244329:G:C	NA	missense	c.1677C>G	p.Cys559Trp

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2:25244329:G:T	NA	stop_gained	c.1677C>A	p.Cys559*
2:25244329:GCA AAAGCA:G	NA	frameshift	c.1669_1676delTG CTTTTG	p.Cys557fs
2:25244329:GCA AAAGCAC:G	NA	inframe_indel	c.1668_1676delGT GCTTTTG	p.Arg556_Cys559 delinsSer
2:25244330:C:A	rs1313738991	missense	c.1676G>T	p.Cys559Phe
2:25244330:C:CA	NA	frameshift	c.1675dupT	p.Cys559fs
2:25244330:C:T	NA	missense	c.1676G>A	p.Cys559Tyr
2:25244330:CA:C	NA	frameshift	c.1675delT	p.Cys559fs
2:25244330:CAA: C	NA	frameshift	c.1674_1675delTT	p.Phe558fs
2:25244331:A:G	NA	missense	c.1675T>C	p.Cys559Arg
2:25244333:A:C	NA	missense	c.1673T>G	p.Phe558Cys
2:25244333:AAG: A	NA	frameshift	c.1671_1672delCT	p.Phe558fs
2:25244335:G:T	NA	stop_gained	c.1671C>A	p.Cys557*
2:25244336:C:T	NA	missense	c.1670G>A	p.Cys557Tyr
2:25244336:CA:C	NA	frameshift	c.1669delT	p.Cys557fs
2:25244336:CACC TGGAA:C	NA	frameshift	c.1668- 6_1669delTTCCAG GT	p.Cys557fs
2:25244337:A:C	rs760791871	missense	c.1669T>G	p.Cys557Gly
2:25244337:A:G	NA	missense	c.1669T>C	p.Cys557Arg
2:25244337:AC:A	rs1267683924	frameshift	c.1668delG	p.Arg556fs
2:25244337:ACC: A	NA	frameshift	c.1668- 1_1668delGG	p.Arg556fs
2:25244338:C:A	NA	missense	c.1668G>T	p.Arg556Ser
2:25244338:C:G	NA	missense	c.1668G>C	p.Arg556Ser
2:25244339:C:A	NA	splice_acceptor	c.1668-1G>T	
2:25244339:C:G	NA	splice_acceptor	c.1668-1G>C	
2:25244339:C:T	NA	splice_acceptor	c.1668-1G>A	

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2:25244339:CT:C	NA	splice_acceptor	c.1668-2delA	
2:25244339:CTGG AA:C	NA	splice_acceptor	c.1668-6_1668- 2delTTCCA	
2:25244340:T:C	rs764270325	splice_acceptor	c.1668-2A>G	
2:25244340:T:G	NA	splice_acceptor	c.1668-2A>C	
2:25244530:ACA GCCTCACCTG:A	NA	splice_donor	c.1665_1667+9del CAGGTGAGGCTG	p.Cys555_Arg556 delinsTrp
2:25244538:A:C	NA	splice_donor	c.1667+2T>G	
2:25244538:A:G	NA	splice_donor	c.1667+2T>C	
2:25244538:A:T	NA	splice_donor	c.1667+2T>A	
2:25244539:C:A	rs776844136	splice_donor	c.1667+1G>T	
2:25244539:C:G	rs776844136	splice_donor	c.1667+1G>C	
2:25244539:C:T	rs776844136	splice_donor	c.1667+1G>A	
2:25244540:C:CT GCA	NA	frameshift	c.1663_1666dupTG CA	p.Arg556fs
2:25244540:C:G	rs1162120698	missense	c.1667G>C	p.Arg556Thr
2:25244540:CT:C	NA	frameshift	c.1666delA	p.Arg556fs
2:25244541:T:A	NA	missense	c.1666A>T	p.Arg556Trp
2:25244541:T:C	rs770311493	missense	c.1666A>G	p.Arg556Gly
2:25244542:G:T	NA	stop_gained	c.1665C>A	p.Cys555*
2:25244543:C:CA	NA	frameshift	c.1663dupT	p.Cys555fs
2:25244543:CA:C	NA	frameshift	c.1663delT	p.Cys555fs
2:25244544:A:G	NA	missense	c.1663T>C	p.Cys555Arg
2:25244545:G:T	NA	stop_gained	c.1662C>A	p.Cys554*
2:25244546:CAG: C	NA	frameshift	c.1659_1660delCT	p.Asn553fs
2:25244547:A:C	NA	missense	c.1660T>G	p.Cys554Gly
2:25244547:A:G	NA	missense	c.1660T>C	p.Cys554Arg
2:25244547:AGT: A	NA	frameshift	c.1658_1659delAC	p.Asn553fs

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2:25244547:AGTT G:A	NA	frameshift	c.1656_1659delCA AC	p.Asn553fs
2:25244548:G:C	NA	missense	c.1659C>G	p.Asn553Lys
2:25244548:G:T	NA	missense	c.1659C>A	p.Asn553Lys
2:25244548:GT:G	NA	frameshift	c.1658delA	p.Asn553fs
2:25244548:GTT: G	NA	frameshift	c.1657_1658delAA	p.Asn553fs
2:25244549:T:TT GTGTTG	NA	frameshift	c.1657_1658insCA ACACA	p.Asn553fs
2:25244550:TG:T	NA	frameshift	c.1656delC	p.Asn552fs
2:25244551:GT:G	rs1229047002	frameshift	c.1655delA	p.Asn552fs
2:25244551:GTTG T:G	NA	frameshift	c.1652_1655delAC AA	p.Asn551fs
2:25244554:G:GT	NA	frameshift	c.1652dupA	p.Asn551fs
2:25244556:TTCC GCACATGAGCACC :T	NA	frameshift	c.1635_1650delGG TGCTCATGTGCGGA	p.Val546fs
2:25244557:TC:T	NA	frameshift	c.1649delG	p.Gly550fs
2:25244557:TCCG :T	NA	stop_gained	c.1647_1649delCG G	p.Cys549_Gly550 delinsTer
2:25244557:TCCG CACATGAGCA:T	NA	frameshift	c.1637_1649delTG CTCATGTGCGG	p.Val546fs
2:25244559:C:A	rs763336898	stop_gained	c.1648G>T	p.Gly550*
2:25244559:C:G	NA	missense	c.1648G>C	p.Gly550Arg
2:25244559:C:T	rs763336898	missense	c.1648G>A	p.Gly550Arg
2:25244559:CGCA CATGA:C	NA	frameshift	c.1640_1647delTC ATGTGC	p.Leu547fs
2:25244560:G:C	NA	missense	c.1647C>G	p.Cys549Trp
2:25244560:G:T	NA	stop_gained	c.1647C>A	p.Cys549*
2:25244561:C:T	NA	missense	c.1646G>A	p.Cys549Tyr
2:25244561:CA:C	NA	frameshift	c.1645delT	p.Cys549fs
2:25244562:A:AC	rs1397787959	frameshift	c.1644dupG	p.Cys549fs
2:25244562:A:AC G	NA	frameshift	c.1644_1645insCG	p.Cys549fs

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2:25244562:A:G	NA	missense	c.1645T>C	p.Cys549Arg
2:25244563:C:A	NA	missense	c.1644G>T	p.Met548Ile
2:25244563:C:T	rs1403215240	missense	c.1644G>A	p.Met548Ile
2:25244563:CAT: C	rs1307594893	frameshift	c.1642_1643delAT	p.Met548fs
2:25244564:A:C	NA	missense	c.1643T>G	p.Met548Arg
2:25244564:A:G	NA	missense	c.1643T>C	p.Met548Thr
2:25244564:A:T	rs587777509	missense	c.1643T>A	p.Met548Lys
2:25244565:T:C	NA	missense	c.1642A>G	p.Met548Val
2:25244566:GAG CACCTCACGGCCC CCACAGCAGATG GTGCAGT:G	NA	frameshift	c.1607_1640delAC TGACCATCTGCTG TGGGGGCCGTGAG GTGCT	p.Tyr536fs
2:25244567:A:C	rs111848085	missense	c.1640T>G	p.Leu547Arg
2:25244567:A:G	rs111848085	missense	c.1640T>C	p.Leu547Pro
2:25244567:A:T	rs111848085	missense	c.1640T>A	p.Leu547His
2:25244568:G:A	rs759176128	missense	c.1639C>T	p.Leu547Phe
2:25244568:G:GA	NA	frameshift	c.1638_1639insT	p.Leu547fs
2:25244570:AC:A	NA	frameshift	c.1636delG	p.Val546fs
2:25244573:T:TC ACGGCCCCACAG CAGATGGTGCAG TAGGA	NA	frameshift	c.1603_1633dupTC CTACTGCACCATCT GCTGTGGGGGCCG TG	p.Glu545fs
2:25244573:TC:T	NA	frameshift	c.1633delG	p.Glu545fs
2:25244574:C:A	NA	stop_gained	c.1633G>T	p.Glu545*
2:25244574:C:CA	NA	frameshift	c.1632dupT	p.Glu545fs
2:25244575:AC:A	NA	frameshift	c.1631delG	p.Arg544fs
2:25244576:CG:C	NA	frameshift	c.1630delC	p.Arg544fs
2:25244577:GGC CCC:G	NA	frameshift	c.1625_1629delGG GGC	p.Gly542fs
2:25244578:G:GC	rs1164367418	frameshift	c.1628dupG	p.Arg544fs
2:25244578:G:GC C	NA	frameshift	c.1627_1628dupG G	p.Arg544fs
2:25244578:GC:G	rs1392385564	frameshift	c.1628delG	p.Gly543fs

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2:25244579:C:A	rs767226511	missense	c.1628G>T	p.Gly543Val
2:25244579:C:G	rs767226511	missense	c.1628G>C	p.Gly543Ala
2:25244579:C:T	rs767226511	missense	c.1628G>A	p.Gly543Asp
2:25244580:C:A	rs752222356	missense	c.1627G>T	p.Gly543Cys
2:25244580:C:T	NA	missense	c.1627G>A	p.Gly543Ser
2:25244582:C:CC ACAGCAGATGGT GCAGTAGGG	NA	frameshift	c.1624_1625insCCC TACTGCACCATCTG CTGTG	p.Gly542fs
2:25244584:A:T	NA	stop_gained	c.1623T>A	p.Cys541*
2:25244585:CA:C	NA	frameshift	c.1621delT	p.Cys541fs
2:25244586:A:AG	NA	frameshift	c.1620dupC	p.Cys541fs
2:25244586:AGC AGATGGTGC:A	NA	frameshift	c.1610_1620delGC ACCATCTGC	p.Cys537fs
2:25244586:AGC AGATGGTGCAGT AG:A	NA	frameshift	c.1605_1620delCT ACTGCACCATCTGC	p.Tyr536fs
2:25244587:G:T	NA	stop_gained	c.1620C>A	p.Cys540*
2:25244588:C:CA	rs1438581688	frameshift	c.1618dupT	p.Cys540fs
2:25244588:CA:C	rs1366761466	frameshift	c.1618delT	p.Cys540fs
2:25244588:CAG AT:C	NA	frameshift	c.1615_1618delAT CT	p.Ile539fs
2:25244588:CAG ATGGTGCAG:C	NA	frameshift	c.1608_1618delCT GCACCATCT	p.Tyr536fs
2:25244589:AG:A	NA	frameshift	c.1617delC	p.Cys540fs
2:25244589:AGAT G:A	NA	frameshift	c.1614_1617delCA TC	p.Ile539fs
2:25244590:GA:G	NA	frameshift	c.1616delT	p.Ile539fs
2:25244592:T:TG	NA	frameshift	c.1614dupC	p.Ile539fs
2:25244592:TG:T	rs1291735877	frameshift	c.1614delC	p.Ile539fs
2:25244592:TGG: T	NA	frameshift	c.1613_1614delCC	p.Thr538fs
2:25244593:G:GG T	NA	frameshift	c.1612_1613dupAC	p.Ile539fs

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2:25244593:G:GT GCAC	NA	frameshift	c.1613_1614insGT GCA	p.Ile539fs
2:25244596:G:C	NA	missense	c.1611C>G	p.Cys537Trp
2:25244596:G:GC A	NA	frameshift	c.1609_1610dupTG	p.Thr538fs
2:25244596:G:T	NA	stop_gained	c.1611C>A	p.Cys537*
2:25244596:GC:G	NA	frameshift	c.1610delG	p.Cys537fs
2:25244597:C:A	NA	missense	c.1610G>T	p.Cys537Phe
2:25244597:C:T	NA	missense	c.1610G>A	p.Cys537Tyr
2:25244598:A:C	NA	missense	c.1609T>G	p.Cys537Gly
2:25244598:A:G	NA	missense	c.1609T>C	p.Cys537Arg
2:25244598:A:T	NA	missense	c.1609T>A	p.Cys537Ser
2:25244598:AG:A	NA	frameshift	c.1608delC	p.Cys537fs
2:25244599:G:C	rs370376334	stop_gained	c.1608C>G	p.Tyr536*
2:25244599:G:GT	NA	frameshift	c.1607dupA	p.Tyr536fs
2:25244599:G:T	NA	stop_gained	c.1608C>A	p.Tyr536*
2:25244599:GT:G	rs759716976	frameshift	c.1607delA	p.Tyr536fs
2:25244600:T:TA GGA	NA	frameshift	c.1603_1606dupTC CT	p.Tyr536fs
2:25244600:TAG: T	NA	frameshift	c.1605_1606delCT	p.Tyr536fs
2:25244601:AG:A	rs767775447	frameshift	c.1605delC	p.Tyr536fs
2:25244601:AGG ACTGGT:A	NA	frameshift	c.1598_1605delAC CAGTCC	p.Tyr533fs
2:25244602:G:GG A	NA	frameshift	c.1603_1604dupTC	p.Tyr536fs
2:25244602:G:GG ACT	NA	frameshift	c.1601_1604dupA GTC	p.Tyr536fs
2:25244604:AC:A	NA	frameshift	c.1602delG	p.Gln534fs
2:25244605:CTGG :C	NA	stop_gained	c.1599_1601delCC A	p.Tyr533_Gln534 delinsTer
2:25244606:TG:T	NA	frameshift	c.1600delC	p.Gln534fs
2:25244607:G:A	rs1167876410	stop_gained	c.1600C>T	p.Gln534*
2:25244608:G:C	rs757057121	stop_gained	c.1599C>G	p.Tyr533*

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2:25244608:G:GT AGCCGTCGTCGTC GTACTGGTA	NA	frameshift	c.1576_1598dupTA CCAGTACGACGACG ACGGCTA	p.Gln534fs
2:25244608:G:T	rs757057121	stop_gained	c.1599C>A	p.Tyr533*
2:25244608:GT:G	NA	frameshift	c.1598delA	p.Tyr533fs
2:25244609:TA:T	NA	frameshift	c.1597delT	p.Tyr533fs
2:25244610:AG:A	NA	frameshift	c.1596delC	p.Tyr533fs
2:25244612:C:A	NA	missense	c.1595G>T	p.Gly532Val
2:25244613:C:CG T	NA	frameshift	c.1592_1593dupAC	p.Gly532fs
2:25244613:C:T	rs951361433	missense	c.1594G>A	p.Gly532Ser
2:25244613:CG:C	rs1329318931	frameshift	c.1593delC	p.Asp531fs
2:25244614:GT:G	NA	frameshift	c.1592delA	p.Asp531fs
2:25244615:TC:T	NA	frameshift	c.1591delG	p.Asp531fs
2:25244616:C:T	rs745721709	missense	c.1591G>A	p.Asp531Asn
2:25244616:CG:C	NA	frameshift	c.1590delC	p.Asp530fs
2:25244617:G:GC	NA	frameshift	c.1589_1590insG	p.Asp530fs
2:25244617:GTCG TCGTA:G	NA	frameshift	c.1582_1589delTA CGACGA	p.Tyr528fs
2:25244621:TCGT A:T	NA	frameshift	c.1582_1585delTA CG	p.Tyr528fs
2:25244621:TCGT ACTGGTACGCACA CTC:T	NA	frameshift	c.1567_1585delGA GTGTGCGTACCAGT ACG	p.Glu523fs
2:25244622:C:CG	NA	frameshift	c.1584dupC	p.Asp529fs
2:25244622:C:T	rs962805778	missense	c.1585G>A	p.Asp529Asn
2:25244623:G:C	NA	stop_gained	c.1584C>G	p.Tyr528*
2:25244623:G:T	NA	stop_gained	c.1584C>A	p.Tyr528*
2:25244624:T:TA CTGGTA	NA	frameshift	c.1576_1582dupTA CCAGT	p.Tyr528fs
2:25244624:TACT GGTA:T	NA	frameshift	c.1576_1582delTA CCAGT	p.Tyr526fs
2:25244625:A:AC	NA	frameshift	c.1581dupG	p.Tyr528fs
2:25244625:AC:A	NA	frameshift	c.1581delG	p.Gln527fs

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2:25244625:ACTG G:A	NA	frameshift	c.1578_1581delCC AG	p.Gln527fs
2:25244626:C:A	NA	missense	c.1581G>T	p.Gln527His
2:25244626:C:G	NA	missense	c.1581G>C	p.Gln527His
2:25244626:CT:C	NA	frameshift	c.1580delA	p.Gln527fs
2:25244627:T:G	rs773701621	missense	c.1580A>C	p.Gln527Pro
2:25244627:TG:T	NA	frameshift	c.1579delC	p.Gln527fs
2:25244628:G:A	rs1372858208	stop_gained	c.1579C>T	p.Gln527*
2:25244628:GGT ACGCACACTCCAG AAAGCAGTTCTA: G	NA	splice_acceptor	c.1555- 3_1578delTAGAAC TGCTTTCTGGAGTG TGCGTAC	p.Asn519_Gln527 del
2:25244629:G:C	rs763384327	stop_gained	c.1578C>G	p.Tyr526*
2:25244629:G:T	NA	stop_gained	c.1578C>A	p.Tyr526*
2:25244629:GTAC GCAC:G	NA	frameshift	c.1571_1577delGT GCGTA	p.Cys524fs
2:25244629:GTAC GCACACTC:G	NA	frameshift	c.1567_1577delGA GTGTGCGTA	p.Glu523fs
2:25244631:AC:A	NA	frameshift	c.1575delG	p.Tyr526fs
2:25244633:GC:G	NA	frameshift	c.1573delG	p.Ala525fs
2:25244633:GCA: G	rs765267873	frameshift	c.1572_1573delTG	p.Ala525fs
2:25244634:C:CA	NA	frameshift	c.1572dupT	p.Ala525fs
2:25244634:CACA CTCCAGA:C	NA	frameshift	c.1563_1572delTCT GGAGTGT	p.Phe521fs
2:25244635:A:T	NA	stop_gained	c.1572T>A	p.Cys524*
2:25244637:AC:A	rs1238915745	frameshift	c.1569delG	p.Glu523fs
2:25244638:CT:C	NA	frameshift	c.1568delA	p.Glu523fs
2:25244639:TC:T	NA	frameshift	c.1567delG	p.Glu523fs
2:25244640:C:A	NA	stop_gained	c.1567G>T	p.Glu523*
2:25244641:CA:C	NA	frameshift	c.1565delT	p.Leu522fs
2:25244643:G:GA	NA	frameshift	c.1563dupT	p.Leu522fs
2:25244643:GA:G	NA	frameshift	c.1563delT	p.Leu522fs

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2:25244647:G:GC A	NA	frameshift	c.1558_1559dupTG	p.Phe521fs
2:25244647:G:T	rs774696234	stop_gained	c.1560C>A	p.Cys520*
2:25244647:GC:G	NA	frameshift	c.1559delG	p.Cys520fs
2:25244648:C:CA	NA	frameshift	c.1558dupT	p.Cys520fs
2:25244648:CA:C	NA	frameshift	c.1558delT	p.Cys520fs
2:25244648:CAGT TCTAGACAGCAGC GGGAAGGGTCAG :C	NA	frameshift	c.1555- 24_1558delCTGAC CCTTCCCGCTGCTG TCTAGAACT	p.Asn519fs
2:25244650:GT:G	NA	frameshift	c.1556delA	p.Asn519fs
2:25244652:T:TC	NA	splice_acceptor	c.1555-1dupG	
2:25244653:C:G	rs759936287	splice_acceptor	c.1555-1G>C	
2:25244653:C:T	NA	splice_acceptor	c.1555-1G>A	
2:25244654:T:A	NA	splice_acceptor	c.1555-2A>T	
2:25244654:T:C	rs767136883	splice_acceptor	c.1555-2A>G	
2:25244654:T:G	rs767136883	splice_acceptor	c.1555-2A>C	
2:25245251:A:C	rs758228404	splice_donor	c.1554+2T>G	
2:25245251:A:G	NA	splice_donor	c.1554+2T>C	
2:25245251:AC:A	NA	splice_donor	c.1554+1delG	
2:25245252:C:A	NA	splice_donor	c.1554+1G>T	
2:25245252:C:G	NA	splice_donor	c.1554+1G>C	
2:25245252:C:T	rs766110518	splice_donor	c.1554+1G>A	
2:25245253:C:CTT GCAGT	NA	frameshift	c.1547_1553dupAC TGCAA	p.Asn519fs
2:25245255:T:A	NA	stop_gained	c.1552A>T	p.Lys518*
2:25245255:T:TG	NA	frameshift	c.1551dupC	p.Lys518fs

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2:25245255:T:TG C	NA	frameshift	c.1550_1551dupGC	p.Lys518fs
2:25245255:TG:T	NA	frameshift	c.1551delC	p.Cys517fs
2:25245256:G:T	NA	stop_gained	c.1551C>A	p.Cys517*
2:25245256:GC:G	NA	frameshift	c.1550delG	p.Cys517fs
2:25245258:AG:A	NA	frameshift	c.1548delC	p.Cys517fs
2:25245259:GT:G	rs746528594	frameshift	c.1547delA	p.Asn516fs
2:25245261:TTTG GCACA:T	NA	frameshift	c.1538_1545delTG TGCCAA	p.Met513fs
2:25245263:T:TG GCACATTCCTCCA ACGAAGAGGGG	NA	frameshift	c.1519_1543dupCC CCTCTTCGTTGGAG GAATGTGCC	p.Gln515fs
2:25245263:TG:T	NA	frameshift	c.1543delC	p.Gln515fs
2:25245263:TGGC ACATTCCTCC:T	NA	frameshift	c.1531_1543delGG AGGAATGTGCC	p.Gly511fs
2:25245263:TGGC ACATTCCTCCAAC GA:T	NA	stop_gained	c.1526_1543delTC GTTGGAGGAATGT GCC	p.Phe509_Gln515 delinsTer
2:25245264:G:A	rs1449820788	stop_gained	c.1543C>T	p.Gln515*
2:25245265:G:T	NA	stop_gained	c.1542C>A	p.Cys514*
2:25245266:CA:C		frameshift	c.1540delT	p.Cys514fs
2:25245267:AC:A	NA	frameshift	c.1539delG	p.Met513fs
2:25245269:A:AT	rs780842310	frameshift	c.1537dupA	p.Met513fs
2:25245269:AT:A	NA	frameshift	c.1537delA	p.Met513fs
2:25245269:ATTC CTCCAACGAAG:A	NA	frameshift	c.1524_1537delCTT CGTTGGAGGAA	p.Phe509fs
2:25245270:TTC:T	NA	frameshift	c.1535_1536delGA	p.Gly512fs
2:25245271:T:TCC TC	NA	frameshift	c.1532_1535dupG AGG	p.Met513fs
2:25245272:C:CC TCCAACGAAGAG GGGGTGTTCAG GGTAACATTGAG	NA	frameshift	c.1498_1534dupCT CAATGTTACCCTGG AACACCCCTCTTC GTTGGAG	p.Gly512fs
2:25245274:TC:T	NA	frameshift	c.1532delG	p.Gly511fs

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2:25245276:C:A	NA	stop_gained	c.1531G>T	p.Gly511*
2:25245276:C:CA	NA	frameshift	c.1530dupT	p.Gly511fs
2:25245276:CA:C	NA	frameshift	c.1530delT	p.Gly511fs
2:25245276:CAAC G:C	NA	frameshift	c.1527_1530delCG TT	p.Phe509fs
2:25245278:AC:A	NA	frameshift	c.1528delG	p.Val510fs
2:25245278:ACG: A	NA	frameshift	c.1527_1528delCG	p.Val510fs
2:25245279:CG:C	NA	frameshift	c.1527delC	p.Phe509fs
2:25245281:AAG: A	NA	frameshift	c.1524_1525delCT	p.Phe509fs
2:25245283:GA:G	NA	frameshift	c.1523delT	p.Leu508fs
2:25245284:A:AG	NA	frameshift	c.1522dupC	p.Leu508fs
2:25245284:A:G	rs1056677010	missense	c.1523T>C	p.Leu508Pro
2:25245284:AG:A	rs1162765172	frameshift	c.1522delC	p.Leu508fs
2:25245288:G:GG TGTT	NA	frameshift	c.1514_1518dupAA CAC	p.Pro507fs
2:25245289:GT:G	NA	frameshift	c.1517delA	p.His506fs
2:25245290:T:TC	NA	frameshift	c.1516_1517insG	p.His506fs
2:25245290:T:TCC	NA	frameshift	c.1516_1517insGG	p.His506fs
2:25245290:TGTT CC:T	NA	frameshift	c.1512_1516delGG AAC	p.Glu505fs
2:25245290:TGTT CCAG:T	NA	frameshift	c.1510_1516delCT GGAAC	p.Leu504fs
2:25245291:GT:G	NA	frameshift	c.1515delA	p.Glu505fs
2:25245293:T:TCC AGGGTAACATTGA GGCTCCC	NA	frameshift	c.1492_1513dupG GGAGCCTCAATGTT ACCCTGG	p.Glu505fs
2:25245293:TC:T	NA	frameshift	c.1513delG	p.Glu505fs
2:25245293:TCCA GGGTAACATTGA GGCTCCCACA:T	NA	frameshift	c.1489_1513delTG TGGGAGCCTCAATG TTACCCTGG	p.Cys497fs
2:25245294:C:A	rs373860660	stop_gained	c.1513G>T	p.Glu505*

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2:25245294:C:CC AGGGTAACATTGA GGCTCCCACAGG	NA	frameshift	c.1487_1512dupCC TGTGGGAGCCTCAA TGTTACCCTG	p.Glu505fs
2:25245295:CA:C	NA	frameshift	c.1511delT	p.Leu504fs
2:25245295:CAG GG:C	NA	frameshift	c.1508_1511delCC CT	p.Thr503fs
2:25245296:AG:A	NA	frameshift	c.1510delC	p.Leu504fs
2:25245298:G:GG T	NA	frameshift	c.1507_1508dupAC	p.Leu504fs
2:25245298:G:GG TAA	NA	frameshift	c.1505_1508dupTT AC	p.Leu504fs
2:25245298:GGT: G	NA	frameshift	c.1507_1508delAC	p.Thr503fs
2:25245299:G:GT	NA	frameshift	c.1507dupA	p.Thr503fs
2:25245299:GT:G	NA	frameshift	c.1507delA	p.Thr503fs
2:25245300:T:TA	NA	frameshift	c.1506dupT	p.Thr503fs
2:25245300:TA:T	NA	frameshift	c.1506delT	p.Thr503fs
2:25245300:TAA: T	NA	frameshift	c.1505_1506delTT	p.Val502fs
2:25245302:AC:A	NA	frameshift	c.1504delG	p.Val502fs
2:25245303:C:CA	NA	frameshift	c.1503dupT	p.Val502fs
2:25245303:CA:C	NA	frameshift	c.1503delT	p.Asn501fs
2:25245303:CAT: C	NA	frameshift	c.1502_1503delAT	p.Asn501fs
2:25245304:A:AT T	NA	frameshift	c.1501_1502dupAA	p.Asn501fs
2:25245304:AT:A	NA	frameshift	c.1502delA	p.Asn501fs
2:25245304:ATT: A	NA	frameshift	c.1501_1502delAA	p.Asn501fs
2:25245304:ATTG AG:A	NA	frameshift	c.1498_1502delCT CAA	p.Leu500fs
2:25245306:TGA: T	NA	frameshift	c.1499_1500delTC	p.Leu500fs
2:25245308:AG:A	NA	frameshift	c.1498delC	p.Leu500fs

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2:25245309:G:GG CTC	NA	frameshift	c.1494_1497dupG AGC	p.Leu500fs
2:25245311:CT:C	NA	frameshift	c.1495delA	p.Ser499fs
2:25245312:TC:T	NA	frameshift	c.1494delG	p.Ser499fs
2:25245314:C:CC A	NA	frameshift	c.1491_1492dupTG	p.Gly498fs
2:25245315:C:CA	NA	frameshift	c.1491dupT	p.Gly498fs
2:25245316:A:C	NA	missense	c.1491T>G	p.Cys497Trp
2:25245316:AC:A	NA	frameshift	c.1490delG	p.Cys497fs
2:25245317:C:A	rs779323387	missense	c.1490G>T	p.Cys497Phe
2:25245317:C:G	NA	missense	c.1490G>C	p.Cys497Ser
2:25245317:C:T	rs779323387	missense	c.1490G>A	p.Cys497Tyr
2:25245317:CA:C	NA	frameshift	c.1489delT	p.Cys497fs
2:25245317:CAG GAGATGCAGATG TCTGGAA:C	NA	splice_acceptor	c.1475- 6_1489delTTCCAG ACATCTGCATCTCCT	p.Asp492_Cys497 delinsGly
2:25245318:A:G	NA	missense	c.1489T>C	p.Cys497Arg
2:25245318:A:T	NA	missense	c.1489T>A	p.Cys497Ser
2:25245318:AG:A	NA	frameshift	c.1488delC	p.Cys497fs
2:25245320:GA:G	NA	frameshift	c.1486delT	p.Ser496fs
2:25245321:AG:A	NA	frameshift	c.1485delC	p.Ser496fs
2:25245325:G:C	rs1224606813	missense	c.1482C>G	p.Cys494Trp
2:25245325:G:T	NA	stop_gained	c.1482C>A	p.Cys494*
2:25245326:C:A	NA	missense	c.1481G>T	p.Cys494Phe
2:25245326:C:G	rs1240736156	missense	c.1481G>C	p.Cys494Ser
2:25245326:C:T	NA	missense	c.1481G>A	p.Cys494Tyr
2:25245327:A:C	NA	missense	c.1480T>G	p.Cys494Gly
2:25245327:A:G	NA	missense	c.1480T>C	p.Cys494Arg
2:25245327:A:T	NA	missense	c.1480T>A	p.Cys494Ser
2:25245327:AG:A	NA	frameshift	c.1479delC	p.Cys494fs
2:25245328:GA:G	NA	frameshift	c.1478delT	p.Ile493fs
2:25245329:AT:A	NA	frameshift	c.1477delA	p.Ile493fs
2:25245330:TG:T	NA	frameshift	c.1476delC	p.Asp492fs

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2:25245330:TGTC TGGAAAGCA:T	NA	frameshift	c.1475- 10_1476delTGCTTT CCAGAC	p.Ile493fs
2:25245331:GT:G	NA	frameshift	c.1475delA	p.Asp492fs
2:25245333:C:G	rs746402248	splice_acceptor	c.1475-1G>C	
2:25245333:C:T	NA	splice_acceptor	c.1475-1G>A	
2:25245333:CT:C	NA	splice_acceptor	c.1475-2delA	
2:25245333:CTGG AAAGCAGAGGGA GGGGA:C	NA	splice_acceptor	c.1475-21_1475- 2delTCCCCTCCCTCT GCTTTCCA	
2:25246010:ACAA ACTTACCCT:A	NA	splice_donor	c.1472_1474+9del AGGGTAAGTTTG	p.Glu491del
2:25246015:CTTA CCCTCAATG:C	NA	frameshift	c.1467_1474+4del CATTGAGGGTAA	p.Asn489fs
2:25246018:A:AC	NA	splice_donor	c.1474+1dupG	
2:25246018:A:C	NA	splice_donor	c.1474+2T>G	
2:25246018:AC:A	rs1200077617	splice_donor	c.1474+1delG	
2:25246018:ACCC T:A	NA	splice_donor	c.1472_1474+1del AGGG	p.Glu491del
2:25246019:C:A	NA	splice_donor	c.1474+1G>T	
2:25246019:C:G	NA	splice_donor	c.1474+1G>C	
2:25246019:C:T	rs761608224	splice_donor	c.1474+1G>A	
2:25246022:T:TC A	NA	frameshift	c.1470_1471dupTG	p.Glu491fs
2:25246022:TC:T	NA	frameshift	c.1471delG	p.Glu491fs
2:25246022:TCAA TGTTCCGG:T	NA	frameshift	c.1461_1471delCC GGAACATTG	p.Cys487fs
2:25246023:C:A	NA	stop_gained	c.1471G>T	p.Glu491*
2:25246023:C:CA	NA	frameshift	c.1470dupT	p.Glu491fs
2:25246023:CA:C	NA	frameshift	c.1470delT	p.Ile490fs

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2:25246024:A:AA TGGTGCGGC	NA	frameshift	c.1469_1470insGC CGCACCAT	p.Ile490fs
2:25246025:A:AT GTTCCGGCACTTC	NA	frameshift	c.1455_1468dupG AAGTGCCGGAACA	p.Ile490fs
2:25246026:TG:T	NA	frameshift	c.1467delC	p.Asn489fs
2:25246027:GT:G	NA	frameshift	c.1466delA	p.Asn489fs
2:25246028:TTCC GGCACTTC:T	NA	frameshift	c.1455_1465delGA AGTGCCGGA	p.Lys486fs
2:25246029:T:TC	NA	frameshift	c.1464dupG	p.Asn489fs
2:25246029:TC:T		frameshift	c.1464delG	p.Asn489fs
2:25246030:CCG GCA:C	NA	frameshift	c.1459_1463delTG CCG	p.Cys487fs
2:25246031:C:CG	NA	frameshift	c.1462dupC	p.Arg488fs
2:25246033:G:T	NA	stop_gained	c.1461C>A	p.Cys487*
2:25246033:GC:G	NA	frameshift	c.1460delG	p.Cys487fs
2:25246034:CA:C	NA	frameshift	c.1459delT	p.Cys487fs
2:25246035:AC:A	NA	frameshift	c.1458delG	p.Lys486fs
2:25246035:ACTT CT:A	NA	frameshift	c.1454_1458delAG AAG	p.Gln485fs
2:25246036:CT:C	NA	frameshift	c.1457delA	p.Lys486fs
2:25246037:TTC:T	NA	frameshift	c.1455_1456delGA	p.Lys486fs
2:25246038:T:A	NA	stop_gained	c.1456A>T	p.Lys486*
2:25246038:TC:T	NA	frameshift	c.1455delG	p.Lys486fs
2:25246040:TGCC GCACCTCG:T	NA	frameshift	c.1443_1453delCG AGGTGCGGC	p.Tyr481fs
2:25246041:G:A	NA	stop_gained	c.1453C>T	p.Gln485*
2:25246041:GC:G	NA	frameshift	c.1452delG	p.Gln485fs
2:25246042:C:CC G	rs1131691760	frameshift	c.1450_1451dupCG	p.Gln485fs
2:25246042:CCG: C	NA	frameshift	c.1450_1451delCG	p.Arg484fs
2:25246044:G:GC	NA	frameshift	c.1449dupG	p.Arg484fs
2:25246044:G:GC A	NA	frameshift	c.1448_1449dupTG	p.Arg484fs

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2:25246045:CACC TCGT:C	NA	frameshift	c.1442_1448delAC GAGGT	p.Tyr481fs
2:25246046:AC:A	NA	frameshift	c.1447delG	p.Val483fs
2:25246046:ACC: A	NA	frameshift	c.1446_1447delGG	p.Glu482fs
2:25246047:CCTC GTACACCAGCCGC T:C	NA	frameshift	c.1430_1446delAG CGGCTGGTGTACGA G	p.Glu477fs
2:25246050:C:A	rs769071767	stop_gained	c.1444G>T	p.Glu482*
2:25246050:CG:C	NA	frameshift	c.1443delC	p.Tyr481fs
2:25246051:G:C	rs145465364	stop_gained	c.1443C>G	p.Tyr481*
2:25246051:G:T	NA	stop_gained	c.1443C>A	p.Tyr481*
2:25246052:T:TA	rs1273047372	frameshift	c.1441dupT	p.Tyr481fs
2:25246052:TAC: T	NA	frameshift	c.1440_1441delGT	p.Tyr481fs
2:25246054:CACC A:C	NA	frameshift	c.1436_1439delITG GT	p.Leu479fs
2:25246055:A:AC	NA	frameshift	c.1438dupG	p.Val480fs
2:25246057:CA:C	NA	frameshift	c.1436delIT	p.Leu479fs
2:25246058:AG:A	NA	frameshift	c.1435delC	p.Leu479fs
2:25246058:AGCC G:A	NA	frameshift	c.1432_1435delCG GC	p.Arg478fs
2:25246058:AGCC GCTCTGCAAG:A	NA	splice_acceptor	c.1430- 7_1435delCTTGCA GAGCGGC	p.Glu477_Leu479 delinsVal
2:25246059:GC:G	NA	frameshift	c.1434delG	p.Leu479fs
2:25246061:CG:C	NA	frameshift	c.1432delC	p.Arg478fs
2:25246061:CGCT CTGCAAGGGGAG GAGAGCT:C	NA	splice_acceptor	c.1430- 19_1432delAGCTC TCCTCCCCTTGCA AGC	p.Glu477_Arg478 delinsGly
2:25246062:G:GC T	NA	frameshift	c.1430_1431dupA G	p.Arg478fs

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2:25246062:G:GC TCT	NA	frameshift	c.1430- 2_1431dupAGAG	p.Leu479fs
2:25246062:GC:G	NA	frameshift	c.1431delG	p.Glu477fs
2:25246062:GCTC :G	NA	frameshift	c.1430- 1_1431delGAG	p.Glu477fs
2:25246064:T:TC	NA	splice_accepto r	c.1430-1dupG	
2:25246065:C:A	rs1363411956	splice_accepto r	c.1430-1G>T	
2:25246065:C:G	NA	splice_accepto r	c.1430-1G>C	
2:25246065:C:T	NA	splice_accepto r	c.1430-1G>A	
2:25246066:T:A	NA	splice_accepto r	c.1430-2A>T	
2:25246066:T:C	rs1307856488	splice_accepto r	c.1430-2A>G	
2:25246066:T:G	NA	splice_accepto r	c.1430-2A>C	
2:25246155:ACTA CCTCTTG:A	NA	splice_donor	c.1424_1429+4del CAAGAGGTAG	p.Thr475_Glu477 delinsLys
2:25246157:TA:T	NA	splice_donor	c.1429+2delT	
2:25246158:A:C	NA	splice_donor	c.1429+2T>G	
2:25246158:A:G	rs1207237998	splice_donor	c.1429+2T>C	
2:25246158:A:T	NA	splice_donor	c.1429+2T>A	
2:25246158:AC:A	rs772195614	splice_donor	c.1429+1delG	
2:25246159:C:A	NA	splice_donor	c.1429+1G>T	
2:25246159:C:CC T	NA	frameshift	c.1428_1429dupA G	p.Arg478fs
2:25246159:C:G	NA	splice_donor	c.1429+1G>C	
2:25246159:C:T	rs774112139	splice_donor	c.1429+1G>A	
2:25246159:CCT: C	NA	frameshift	c.1428_1429delAG	p.Glu477fs

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2:25246159:CCTC TTGT:C	NA	frameshift	c.1423_1429delAC AAGAG	p.Thr475fs
2:25246159:CCTC TTGTG:C	NA	frameshift	c.1422_1429delCA CAAGAG	p.Thr475fs
2:25246160:C:A	NA	stop_gained	c.1429G>T	p.Glu477*
2:25246160:CT:C	NA	frameshift	c.1428delA	p.Glu477fs
2:25246162:C:CT	NA	frameshift	c.1426dupA	p.Arg476fs
2:25246162:CT:C	NA	frameshift	c.1426delA	p.Arg476fs
2:25246163:T:A	rs772041976	stop_gained	c.1426A>T	p.Arg476*
2:25246163:TTG: T	NA	frameshift	c.1424_1425delCA	p.Thr475fs
2:25246164:TG:T	NA	frameshift	c.1424delC	p.Thr475fs
2:25246165:G:GT GCGC	NA	frameshift	c.1419_1423dupGC GCA	p.Thr475fs
2:25246165:G:GT GCGCTT	NA	frameshift	c.1423_1424insAA GCGCA	p.Thr475fs
2:25246166:TGC: T	NA	frameshift	c.1421_1422delGC	p.Arg474fs
2:25246167:GC:G	NA	frameshift	c.1421delG	p.Arg474fs
2:25246167:GCG CT:G	NA	frameshift	c.1418_1421delAG CG	p.Glu473fs
2:25246168:C:CT	NA	frameshift	c.1420_1421insA	p.Arg474fs
2:25246168:CG:C	NA	frameshift	c.1420delC	p.Arg474fs
2:25246169:G:GC TCA	NA	frameshift	c.1416_1419dupTG AG	p.Arg474fs
2:25246170:CT:C	NA	frameshift	c.1418delA	p.Glu473fs
2:25246171:TC:T	NA	frameshift	c.1417delG	p.Glu473fs
2:25246172:C:A	NA	stop_gained	c.1417G>T	p.Glu473*
2:25246172:C:CA	NA	frameshift	c.1416dupT	p.Glu473fs
2:25246172:CA:C	NA	frameshift	c.1416delT	p.Asp472fs
2:25246172:CATC A:C	NA	frameshift	c.1413_1416delTG AT	p.Ile471fs
2:25246172:CATC AATAA:C	NA	frameshift	c.1409_1416delTT ATTGAT	p.Ile470fs

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2:25246174:T:TA	NA	frameshift	c.1414_1415insT	p.Asp472fs
2:25246174:TC:T	rs1347154070	frameshift	c.1414delG	p.Asp472fs
2:25246175:CA:C	NA	frameshift	c.1413delT	p.Ile471fs
2:25246178:T:TA	NA	frameshift	c.1410dupT	p.Ile471fs
2:25246178:T:TA A	NA	frameshift	c.1409_1410dupTT	p.Ile471fs
2:25246178:T:TA ATC	NA	frameshift	c.1407_1410dupG ATT	p.Ile471fs
2:25246178:T:TA ATCTCCTTG	NA	frameshift	c.1401_1410dupCA AGGAGATT	p.Ile471fs
2:25246178:TA:T	NA	frameshift	c.1410delT	p.Ile471fs
2:25246179:A:AT	NA	frameshift	c.1409_1410insA	p.Ile471fs
2:25246179:AAT: A	NA	frameshift	c.1408_1409delAT	p.Ile470fs
2:25246180:A:AT C	NA	frameshift	c.1407_1408dupG A	p.Ile470fs
2:25246180:AT:A	rs1348892100	frameshift	c.1408delA	p.Ile470fs
2:25246180:ATC: A	NA	frameshift	c.1407_1408delGA	p.Glu469fs
2:25246180:ATCT CCTTGACCTTGGG C:A	NA	frameshift	c.1392_1408delGC CCAAGGTCAAGGA GA	p.Lys464fs
2:25246182:CT:C	NA	frameshift	c.1406delA	p.Glu469fs
2:25246182:CTCC TTGACCT:C	NA	frameshift	c.1397_1406delAG GTCAAGGA	p.Lys466fs
2:25246183:TC:T	NA	frameshift	c.1405delG	p.Glu469fs
2:25246184:C:A	rs765389013	stop_gained	c.1405G>T	p.Glu469*
2:25246185:C:CT	NA	frameshift	c.1403dupA	p.Glu469fs
2:25246185:CT:C	NA	frameshift	c.1403delA	p.Lys468fs
2:25246187:TG:T	NA	frameshift	c.1401delC	p.Lys468fs
2:25246189:AC:A	rs1360596024	frameshift	c.1399delG	p.Val467fs
2:25246189:ACC: A	NA	frameshift	c.1398_1399delGG	p.Lys466fs
2:25246191:CT:C	rs775785710	frameshift	c.1397delA	p.Lys466fs

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2:25246191:CTTG GGCT:C	NA	frameshift	c.1391_1397delAG CCCAA	p.Lys464fs
2:25246192:T:TT GGGCTTCTCCGC	NA	frameshift	c.1384_1396dupGC GGAGAAGCCCA	p.Lys466fs
2:25246193:T:A	NA	stop_gained	c.1396A>T	p.Lys466*
2:25246193:T:TG	NA	frameshift	c.1395dupC	p.Lys466fs
2:25246193:TG:T	NA	frameshift	c.1395delC	p.Lys466fs
2:25246196:GCTT CT:G	NA	frameshift	c.1388_1392delAG AAG	p.Glu463fs
2:25246197:C:CG AGAA	NA	frameshift	c.1391_1392insTTC TC	p.Lys464fs
2:25246198:T:TTC	NA	frameshift	c.1389_1390dupG A	p.Lys464fs
2:25246198:TTC:T	rs1349621546	frameshift	c.1389_1390delGA	p.Lys464fs
2:25246199:T:A	NA	stop_gained	c.1390A>T	p.Lys464*
2:25246201:T:TC	NA	frameshift	c.1387dupG	p.Glu463fs
2:25246201:TC:T	NA	frameshift	c.1387delG	p.Glu463fs
2:25246201:TCCG CTGTG:T	NA	frameshift	c.1380_1387delCA CAGCGG	p.Ser460fs
2:25246202:C:A	NA	stop_gained	c.1387G>T	p.Glu463*
2:25246202:C:CA	NA	frameshift	c.1386_1387insT	p.Glu463fs
2:25246203:CG:C	NA	frameshift	c.1385delC	p.Ala462fs
2:25246205:C:CT GTGCTCTTCCGGG GCTTTT	NA	frameshift	c.1364_1383dupAA AAGCCCCGGAAGA GCACA	p.Ala462fs
2:25246205:CT:C	NA	frameshift	c.1383delA	p.Ala462fs
2:25246207:G:GC	NA	frameshift	c.1381_1382insG	p.Thr461fs
2:25246208:TG:T	NA	frameshift	c.1380delC	p.Ser460fs
2:25246209:GCT: G	NA	frameshift	c.1378_1379delAG	p.Ser460fs
2:25246209:GCTC TT:G	NA	frameshift	c.1375_1379delAA GAG	p.Lys459fs

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2:25246211:T:TCT TCCGGGGCTTTTT GG	NA	frameshift	c.1361_1377dupCC AAAAAGCCCCGGA AG	p.Ser460fs
2:25246212:CT:C	NA	frameshift	c.1376delA	p.Lys459fs
2:25246213:TTCC GGGGC:T	NA	frameshift	c.1368_1375delGC CCCGGA	p.Pro457fs
2:25246214:T:A	rs768204905	stop_gained	c.1375A>T	p.Lys459*
2:25246214:T:TC	NA	frameshift	c.1374dupG	p.Lys459fs
2:25246214:TC:T	rs886041641	frameshift	c.1374delG	p.Lys459fs
2:25246214:TCC:T	NA	frameshift	c.1373_1374delGG	p.Arg458fs
2:25246216:C:CG	NA	frameshift	c.1372dupC	p.Arg458fs
2:25246216:CG:C	NA	frameshift	c.1372delC	p.Arg458fs
2:25246219:GGCT TTTTGGCT:G	NA	frameshift	c.1359_1369delAG CCAAAAAGC	p.Ala454fs
2:25246220:GC:G	NA	frameshift	c.1368delG	p.Lys456fs
2:25246221:C:CT	NA	frameshift	c.1367dupA	p.Pro457fs
2:25246226:T:A	NA	stop_gained	c.1363A>T	p.Lys455*
2:25246226:TG:T	rs765073748	frameshift	c.1362delC	p.Lys456fs
2:25246228:G:GC	NA	frameshift	c.1360dupG	p.Ala454fs
2:25246228:GC:G	NA	frameshift	c.1360delG	p.Ala454fs
2:25246230:T:TG G	NA	frameshift	c.1357_1358dupCC	p.Ala454fs
2:25246232:G:GT	NA	frameshift	c.1356dupA	p.Pro453fs
2:25246233:T:TG	NA	frameshift	c.1355dupC	p.Pro453fs
2:25246235:GA:G	NA	frameshift	c.1353delT	p.Pro452fs
2:25246235:GAG GTGGTGCGT:G	NA	frameshift	c.1343_1353delAC GCACCACCT	p.Tyr448fs
2:25246236:AG:A	NA	frameshift	c.1352delC	p.Pro451fs
2:25246236:AGG TGGTGCGTAGG:A	NA	frameshift	c.1340_1352delCC TACGCACCACC	p.Ala447fs
2:25246236:AGG TGGTGCGTAGGC AGCTGCCTCAG:A	NA	frameshift	c.1328_1352delCT GAGGCAGCTGCCTA CGCACCACC	p.Pro443fs
2:25246238:G:GT	NA	frameshift	c.1350dupA	p.Pro451fs

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2:25246238:GT:G	NA	frameshift	c.1350delA	p.Pro451fs
2:25246239:TG:T	rs1287033864	frameshift	c.1349delC	p.Pro450fs
2:25246241:GT:G	NA	frameshift	c.1347delA	p.Pro450fs
2:25246242:T:TG	NA	frameshift	c.1346dupC	p.Pro450fs
2:25246242:TGC: T	NA	frameshift	c.1345_1346delGC	p.Ala449fs
2:25246244:CGTA GGCA:C	NA	frameshift	c.1338_1344delTG CCTAC	p.Ala447fs
2:25246245:G:C	NA	stop_gained	c.1344C>G	p.Tyr448*
2:25246245:G:GT T	NA	frameshift	c.1343_1344insAA	p.Tyr448fs
2:25246245:G:T	rs137931376	stop_gained	c.1344C>A	p.Tyr448*
2:25246246:TA:T	NA	frameshift	c.1342delT	p.Tyr448fs
2:25246246:TAG GCAGCTGCCTC:T	NA	frameshift	c.1330_1342delGA GGCAGCTGCCT	p.Glu444fs
2:25246247:AG:A	NA	frameshift	c.1341delC	p.Tyr448fs
2:25246247:AGG CAGCT:A	NA	frameshift	c.1335_1341delAG CTGCC	p.Ala446fs
2:25246249:GC:G	NA	frameshift	c.1339delG	p.Ala447fs
2:25246249:GCA GCTGCCTCAGGT: G	NA	frameshift	c.1326_1339delAC CTGAGGCAGCTG	p.Glu442fs
2:25246250:C:CA GCT	NA	frameshift	c.1335_1338dupA GCT	p.Ala447fs
2:25246252:GCTG C:G	NA	frameshift	c.1333_1336delGC AG	p.Ala445fs
2:25246252:GCTG CCTCAGGTTCCAC C:G	NA	frameshift	c.1320_1336delGG TGGAACCTGAGGC AG	p.Trp440fs
2:25246253:CTGC CTCAGGTTCCACC CACA:C	NA	frameshift	c.1316_1335delTG TGGGTGGAACCTG AGGCA	p.Met439fs
2:25246254:TG:T	NA	frameshift	c.1334delC	p.Ala445fs
2:25246259:C:A	rs1481248882	stop_gained	c.1330G>T	p.Glu444*

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2:25246259:CA:C	NA	frameshift	c.1329delT	p.Glu444fs
2:25246259:CAG: C	NA	frameshift	c.1328_1329delCT	p.Pro443fs
2:25246260:A:AG	NA	frameshift	c.1328dupC	p.Glu444fs
2:25246260:AG:A	NA	frameshift	c.1328delC	p.Pro443fs
2:25246262:GT:G	NA	frameshift	c.1326delA	p.Glu442fs
2:25246265:C:A	rs1205125035	stop_gained	c.1324G>T	p.Glu442*
2:25246266:CA:C	NA	frameshift	c.1322delT	p.Val441fs
2:25246267:A:AC	NA	frameshift	c.1321dupG	p.Val441fs
2:25246267:AC:A	rs1440952501	frameshift	c.1321delG	p.Val441fs
2:25246269:C:CC A	NA	frameshift	c.1318_1319dupTG	p.Trp440fs
2:25246269:C:T	rs866490834	stop_gained	c.1320G>A	p.Trp440*
2:25246270:C:T	rs773260349	stop_gained	c.1319G>A	p.Trp440*
2:25246270:CA:C	NA	frameshift	c.1318delT	p.Trp440fs
2:25246271:A:AC	NA	frameshift	c.1317dupG	p.Trp440fs
2:25246271:AC:A	NA	frameshift	c.1317delG	p.Met439fs
2:25246272:CAT: C	rs762868483	frameshift	c.1315_1316delAT	p.Met439fs
2:25246272:CATG TCCGTGT:C	NA	frameshift	c.1307_1316delAC ACGGACAT	p.Tyr436fs
2:25246273:A:AA T	NA	frameshift	c.1315_1316insAT	p.Met439fs
2:25246273:A:AT G	NA	frameshift	c.1314_1315dupCA	p.Met439fs
2:25246273:AT:A	NA	frameshift	c.1315delA	p.Met439fs
2:25246274:T:TG TCC	NA	frameshift	c.1311_1314dupG GAC	p.Met439fs
2:25246274:TGTC C:T	NA	frameshift	c.1311_1314delGG AC	p.Asp438fs
2:25246275:G:GT CCGTGTACACTTC TTTGTA	NA	frameshift	c.1294_1313dupTA CAAAGAAGGTGTACA CGGA	p.Met439fs
2:25246276:T:TC	NA	frameshift	c.1312dupG	p.Asp438fs

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2:25246276:TC:T		frameshift	c.1312delG	p.Asp438fs
2:25246276:TCCG TGTACACTTCTTT GTA:T	NA	frameshift	c.1294_1312delTA CAAAGAAGTGTACA CGG	p.Tyr432fs
2:25246279:GT:G	NA	frameshift	c.1309delA	p.Thr437fs
2:25246281:G:C	NA	stop_gained	c.1308C>G	p.Tyr436*
2:25246281:G:T	NA	stop_gained	c.1308C>A	p.Tyr436*
2:25246282:T:TA	NA	frameshift	c.1306dupT	p.Tyr436fs
2:25246282:TA:T	NA	frameshift	c.1306delT	p.Tyr436fs
2:25246282:TAC: T	NA	frameshift	c.1305_1306delGT	p.Tyr436fs
2:25246283:ACAC TT:A	NA	frameshift	c.1301_1305delAA GTG	p.Glu434fs
2:25246283:ACAC TTCTTTGT:A	NA	frameshift	c.1295_1305delAC AAAGAAGTG	p.Tyr432fs
2:25246284:CACT TCTTTGTAGGG:C	NA	frameshift	c.1291_1304delCC CTACAAAGAAGT	p.Pro431fs
2:25246285:A:AC	NA	frameshift	c.1303dupG	p.Val435fs
2:25246285:A:AC T	NA	frameshift	c.1302_1303dupA G	p.Val435fs
2:25246286:CT:C	NA	frameshift	c.1302delA	p.Val435fs
2:25246287:T:TTC TTTGTA	NA	frameshift	c.1294_1301dupTA CAAAGA	p.Glu434fs
2:25246287:TTC:T	NA	frameshift	c.1300_1301delGA	p.Glu434fs
2:25246288:TC:T	NA	frameshift	c.1300delG	p.Glu434fs
2:25246289:C:A	NA	stop_gained	c.1300G>T	p.Glu434*
2:25246289:CT:C	NA	frameshift	c.1299delA	p.Glu434fs
2:25246291:T:TT G	NA	frameshift	c.1296_1297dupCA	p.Lys433fs
2:25246292:T:A	NA	stop_gained	c.1297A>T	p.Lys433*
2:25246292:T:TG TAG	NA	frameshift	c.1293_1296dupCT AC	p.Lys433fs
2:25246292:TGTA G:T	NA	frameshift	c.1293_1296delCT AC	p.Tyr432fs

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2:25246293:G:C	NA	stop_gained	c.1296C>G	p.Tyr432*
2:25246293:G:GT AGGGATTTC	NA	stop_gained	c.1287_1295dupG AATCCCTA	p.Pro431_Tyr432i nsTerAsnPro
2:25246293:G:T	NA	stop_gained	c.1296C>A	p.Tyr432*
2:25246294:TA:T	NA	frameshift	c.1294delT	p.Tyr432fs
2:25246295:AG:A	NA	frameshift	c.1293delC	p.Tyr432fs
2:25246295:AGG: A	NA	frameshift	c.1292_1293delCC	p.Pro431fs
2:25246297:G:GC	NA	frameshift	c.1291_1292insG	p.Pro431fs
2:25246297:G:GG A	NA	frameshift	c.1290_1291dupTC	p.Pro431fs
2:25246298:G:GA	NA	frameshift	c.1290dupT	p.Pro431fs
2:25246298:GA:G	NA	frameshift	c.1290delT	p.Tyr432fs
2:25246300:T:TTC TTC	NA	frameshift	c.1284_1288dupG AAGA	p.Asn430fs
2:25246300:TTCT TC:T	NA	frameshift	c.1284_1288delGA AGA	p.Lys429fs
2:25246303:TTC:T	NA	frameshift	c.1284_1285delGA	p.Lys429fs
2:25246304:T:A	NA	stop_gained	c.1285A>T	p.Lys429*
2:25246304:TCTC TTCTGG:T	NA	frameshift	c.1280- 4_1284delCCAGAA GAG	p.Lys429fs
2:25246305:CT:C	rs751049400	frameshift	c.1283delA	p.Glu428fs
2:25246306:TC:T	NA	frameshift	c.1282delG	p.Glu428fs
2:25246307:C:A	NA	stop_gained	c.1282G>T	p.Glu428*
2:25246307:C:CT	NA	frameshift	c.1281dupA	p.Glu428fs
2:25246307:CT:C	NA	frameshift	c.1281delA	p.Glu428fs
2:25246618:A:G	NA	splice_donor	c.1279+2T>C	
2:25246619:C:A	NA	splice_donor	c.1279+1G>T	
2:25246619:C:G	NA	splice_donor	c.1279+1G>C	
2:25246619:C:T	rs374440649	splice_donor	c.1279+1G>A	
2:25246620:C:A	NA	stop_gained	c.1279G>T	p.Glu427*
2:25246620:CT:C	NA	frameshift	c.1278delA	p.Glu427fs

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2:25246623:C:A	NA	stop_gained	c.1276G>T	p.Glu426*
2:25246623:CT:C	NA	frameshift	c.1275delA	p.Glu426fs
2:25246623:CTG:C	NA	frameshift	c.1274_1275delCA	p.Pro425fs
2:25246624:TG:T	NA	frameshift	c.1274delC	p.Pro425fs
2:25246630:CT:C	NA	frameshift	c.1268delA	p.Glu423fs
2:25246632:C:A	NA	stop_gained	c.1267G>T	p.Glu423*
2:25246632:C:CC A	NA	frameshift	c.1265_1266dupTG	p.Glu423fs
2:25246632:C:CT AGG	NA	frameshift	c.1266_1267insCCT A	p.Glu423fs
2:25246633:CAG GCCCTTAG:C	NA	frameshift	c.1256_1265delCT AAGGGCCT	p.Pro419fs
2:25246634:AG:A	rs752298245	frameshift	c.1264delC	p.Leu422fs
2:25246636:GC:G	NA	frameshift	c.1262delG	p.Gly421fs
2:25246636:GCCC T:G	NA	frameshift	c.1259_1262delAG GG	p.Lys420fs
2:25246637:CCCT TAGGGCCAGAAG GCTGGAAGCCCC CAGGGCCATTCA AT:C	NA	frameshift	c.1219_1261delAT TGAATGGGCCCTGG GGGGCTTCCAGCCT TCTGGCCCTAAGG	p.Ile407fs
2:25246639:CT:C	NA	frameshift	c.1259delA	p.Lys420fs
2:25246640:TTAG GGCCAG:T	NA	stop_gained	c.1250_1258delCT GGCCCTA	p.Ser417_Lys420 delinsTer
2:25246640:TTAG GGCCAGA:T	NA	frameshift	c.1249_1258delTCT GGCCCTA	p.Ser417fs
2:25246641:T:A	NA	stop_gained	c.1258A>T	p.Lys420*
2:25246641:T:TA	NA	frameshift	c.1257dupT	p.Lys420fs
2:25246641:TA:T	NA	frameshift	c.1257delT	p.Lys420fs
2:25246642:A:AG	NA	frameshift	c.1256dupC	p.Lys420fs
2:25246642:AG:A	NA	frameshift	c.1256delC	p.Pro419fs
2:25246645:GC:G	NA	frameshift	c.1253delG	p.Gly418fs

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2:25246645:GCCA GAAGGCTGGAAG CCCC:G	NA	frameshift	c.1235_1253delGG GGCTTCCAGCCTTC TGG	p.Gly412fs
2:25246647:C:CA	NA	frameshift	c.1251dupT	p.Gly418fs
2:25246647:CAG: C	NA	frameshift	c.1250_1251delCT	p.Ser417fs
2:25246648:A:AG CCTTCTGGCCCTA AGGGCCT	NA	stop_gained	c.1250_1251insAG GCCCTTAGGGCCAG AAGGC	p.Ser417_Gly418i nsGlyProTerGlyGln LysAla
2:25246648:AG:A	NA	frameshift	c.1250delC	p.Ser417fs
2:25246649:G:GT	NA	frameshift	c.1249_1250insA	p.Ser417fs
2:25246649:GA:G	NA	frameshift	c.1249delT	p.Ser417fs
2:25246651:AG:A	NA	frameshift	c.1247delC	p.Pro416fs
2:25246653:GC:G	NA	frameshift	c.1245delG	p.Gln415fs
2:25246653:GCTG GAAGCCCCCAG GGCCATT:G	NA	frameshift	c.1223_1245delAA TGGGCCCTGGGGG GCTTCCAG	p.Glu408fs
2:25246654:C:CT	NA	frameshift	c.1244dupA	p.Pro416fs
2:25246654:CTGG AAGCCCCCAGG GCCCATTCATCA: C	NA	frameshift	c.1217_1244delTG ATTGAATGGGCCCT GGGGGGCTTCCA	p.Met406fs
2:25246655:TG:T	NA	frameshift	c.1243delC	p.Gln415fs
2:25246656:G:A	rs754223052	stop_gained	c.1243C>T	p.Gln415*
2:25246656:G:GG A	NA	frameshift	c.1241_1242dupTC	p.Gln415fs
2:25246657:G:C	NA	missense	c.1242C>G	p.Phe414Leu
2:25246657:GA:G	NA	frameshift	c.1241delT	p.Phe414fs
2:25246657:GAA GCCCCCAGGGCC CATTC:G	NA	frameshift	c.1222_1241delGA ATGGGCCCTGGGG GGCTT	p.Glu408fs
2:25246658:A:C	NA	missense	c.1241T>G	p.Phe414Cys
2:25246658:A:G	rs1258189576	missense	c.1241T>C	p.Phe414Ser

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2:25246658:AAG CCCCC:A	NA	frameshift	c.1233_1240delGG GGGGCT	p.Gly412fs
2:25246658:AAG CCCCCAGGGCC: A	NA	frameshift	c.1227_1240delGG CCCTGGGGGGCT	p.Trp409fs
2:25246659:A:C	NA	missense	c.1240T>G	p.Phe414Val
2:25246659:A:G	NA	missense	c.1240T>C	p.Phe414Leu
2:25246659:A:T	rs1195155939	missense	c.1240T>A	p.Phe414Ile
2:25246659:AG:A		frameshift	c.1239delC	p.Phe414fs
2:25246659:AGC: A	NA	frameshift	c.1238_1239delGC	p.Gly413fs
2:25246660:G:GC	rs1234388246	frameshift	c.1238dupG	p.Phe414fs
2:25246660:G:GC CCCC	NA	frameshift	c.1234_1238dupG GGGG	p.Phe414fs
2:25246660:GC:G	rs755744291	frameshift	c.1238delG	p.Gly413fs
2:25246663:C:CC CCA	NA	frameshift	c.1232_1235dupTG GG	p.Phe414fs
2:25246667:A:AG	NA	frameshift	c.1231dupC	p.Leu411fs
2:25246667:A:AG GGCCATTCAATC ATGGGCTTG	NA	frameshift	c.1209_1231dupCA AGCCCATGATTGAA TGGGCC	p.Leu411fs
2:25246667:AG:A	NA	frameshift	c.1231delC	p.Leu411fs
2:25246670:GC:G	NA	frameshift	c.1228delG	p.Ala410fs
2:25246672:C:T		stop_gained	c.1227G>A	p.Trp409*
2:25246672:CCAT T:C	rs778152015	frameshift	c.1223_1226delAA TG	p.Glu408fs
2:25246673:C:CA	NA	frameshift	c.1225dupT	p.Trp409fs
2:25246673:C:T	NA	stop_gained	c.1226G>A	p.Trp409*
2:25246674:AT:A	NA	frameshift	c.1224delA	p.Glu408fs
2:25246676:T:TC	NA	frameshift	c.1222dupG	p.Glu408fs
2:25246677:C:A	NA	stop_gained	c.1222G>T	p.Glu408*
2:25246677:C:CA	NA	frameshift	c.1221dupT	p.Glu408fs
2:25246679:A:AT GGGCC	NA	frameshift	c.1219_1220insGG GCCCA	p.Ile407fs

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2:25246681:C:CA A	NA	frameshift	c.1217_1218insTT	p.Met406fs
2:25246681:CATG GGCTTGTT:C	NA	frameshift	c.1207_1217delAA CAAGCCCAT	p.Asn403fs
2:25246683:TG:T	NA	frameshift	c.1215delC	p.Met406fs
2:25246686:GC:G	NA	frameshift	c.1212delG	p.Lys404fs
2:25246687:C:CT	NA	frameshift	c.1211dupA	p.Pro405fs
2:25246689:T:A	NA	stop_gained	c.1210A>T	p.Lys404*
2:25246689:T:TG		frameshift	c.1209dupC	p.Lys404fs
2:25246689:TG:T	rs771398281	frameshift	c.1209delC	p.Asn403fs
2:25246690:GT:G	NA	frameshift	c.1208delA	p.Asn403fs
2:25246691:T:TTC TGACCTCCACGG CCTTGGCAGTGTC ACTCTCATCGCTG	NA	frameshift	c.1167_1207dupCA GCGATGAGAGTGA CACTGCCAAGGCCG TGGAGGTGCAGA	p.Asn403fs
2:25246691:TTCT GCACCTCCACGGC CTTG:T	NA	frameshift	c.1188_1207delCA AGGCCGTGGAGGT GCAGA	p.Lys397fs
2:25246692:TC:T	NA	frameshift	c.1206delG	p.Asn403fs
2:25246693:CT:C	NA	frameshift	c.1205delA	p.Gln402fs
2:25246694:TGCA CCTCCACGGCCTT G:T	NA	frameshift	c.1188_1204delCA AGGCCGTGGAGGT GC	p.Lys397fs
2:25246695:G:A	rs796065342	stop_gained	c.1204C>T	p.Gln402*
2:25246696:C:CA CCT	NA	frameshift	c.1199_1202dupA GGT	p.Gln402fs
2:25246699:CT:C	NA	frameshift	c.1199delA	p.Glu400fs
2:25246699:CTCC ACGGCCTT:C	NA	frameshift	c.1189_1199delAA GGCCGTGGA	p.Lys397fs
2:25246700:T:TC	NA	frameshift	c.1198dupG	p.Glu400fs
2:25246700:TC:T	NA	frameshift	c.1198delG	p.Glu400fs
2:25246700:TCC:T	NA	frameshift	c.1197_1198delGG	p.Glu400fs
2:25246701:C:A	rs751109858	stop_gained	c.1198G>T	p.Glu400*

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2:25246701:C:CC A	NA	frameshift	c.1196_1197dupTG	p.Glu400fs
2:25246701:CCAC GGCCT:C	NA	frameshift	c.1190_1197delAG GCCGTG	p.Lys397fs
2:25246703:AC:A	NA	frameshift	c.1195delG	p.Val399fs
2:25246703:ACG GCCTTG:A	NA	frameshift	c.1188_1195delCA AGGCCG	p.Lys397fs
2:25246704:C:CG TGG	NA	frameshift	c.1194_1195insCC AC	p.Val399fs
2:25246704:CG:C	NA	frameshift	c.1194delC	p.Val399fs
2:25246705:GGC CTTGGCAGTGTCA CTCTCATC:G	NA	frameshift	c.1171_1193delGA TGAGAGTGACACTG CCAAGGC	p.Asp391fs
2:25246706:GC:G	NA	frameshift	c.1192delG	p.Ala398fs
2:25246706:GCCT T:G	NA	frameshift	c.1189_1192delAA GG	p.Lys397fs
2:25246710:T:A	NA	stop_gained	c.1189A>T	p.Lys397*
2:25246710:TG:T	NA	frameshift	c.1188delC	p.Lys397fs
2:25246710:TGGC A:T	NA	frameshift	c.1185_1188delTG CC	p.Ala396fs
2:25246712:GC:G	NA	frameshift	c.1186delG	p.Ala396fs
2:25246713:C:CA	rs1462007191	frameshift	c.1185dupT	p.Ala396fs
2:25246714:A:AG T	NA	frameshift	c.1183_1184dupAC	p.Ala396fs
2:25246714:AG:A	NA	frameshift	c.1184delC	p.Thr395fs
2:25246714:AGT: A	NA	frameshift	c.1183_1184delAC	p.Thr395fs
2:25246715:G:GT	NA	frameshift	c.1183dupA	p.Thr395fs
2:25246717:G:GT	NA	frameshift	c.1181dupA	p.Asp394fs
2:25246718:T:TC ACTCTCATCGCTC	NA	frameshift	c.1180_1181insGA GCGATGAGAGTG	p.Asp394fs
2:25246718:T:TC ACTCTCATCGCTG TCGTGG	NA	frameshift	c.1161_1180dupCC ACGACAGCGATGA GAGTG	p.Asp394fs

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2:25246718:T:TC ACTCTCATCGCTG TCGTGGCACACCG GGA CTGC	NA	frameshift	c.1180_1181insGC AGTCCCGGTGTGCC ACGACAGCGATGA GAGTG	p.Asp394fs
2:25246718:TCA: T	NA	frameshift	c.1179_1180delTG	p.Ser393fs
2:25246719:C:CA	NA	frameshift	c.1179dupT	p.Asp394fs
2:25246719:CA:C	rs1224468033	frameshift	c.1179delT	p.Ser393fs
2:25246720:ACT: A	NA	frameshift	c.1177_1178delAG	p.Ser393fs
2:25246721:C:CG A	NA	frameshift	c.1177_1178insTC	p.Ser393fs
2:25246721:C:CT	NA	frameshift	c.1177dupA	p.Ser393fs
2:25246721:CT:C	NA	frameshift	c.1177delA	p.Ser393fs
2:25246722:TCTC ATCG:T	NA	frameshift	c.1170_1176delCG ATGAG	p.Ser390fs
2:25246723:C:CT	NA	frameshift	c.1175dupA	p.Ser393fs
2:25246724:T:TC ATCGCG	NA	frameshift	c.1174_1175insCG CGATG	p.Glu392fs
2:25246725:C:A	rs1472367159	stop_gained	c.1174G>T	p.Glu392*
2:25246726:AT:A	NA	frameshift	c.1172delA	p.Asp391fs
2:25246730:C:CT G	NA	frameshift	c.1167_1168dupCA	p.Ser390fs
2:25246730:C:G	NA	missense	c.1169G>C	p.Ser390Thr
2:25246731:TG:T	NA	frameshift	c.1167delC	p.Asp389fs
2:25246732:G:GT	NA	frameshift	c.1166dupA	p.Asp389fs
2:25246733:TCGT GGCACACCGGGA A:T	NA	frameshift	c.1150_1165delTTC CCGGTGTGCCACG	p.Phe384fs
2:25246735:G:GT	NA	frameshift	c.1163dupA	p.His388fs
2:25246736:TG:T	NA	frameshift	c.1162delC	p.His388fs
2:25246738:G:T	rs1364266797	stop_gained	c.1161C>A	p.Cys387*
2:25246740:AC:A	NA	frameshift	c.1158delG	p.Cys387fs

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2:25246740:ACAC CGGGAACAGCT:A	NA	frameshift	c.1145_1158delAG CTGTTCCCGGTG	p.Lys382fs
2:25246742:A:AC	NA	frameshift	c.1156dupG	p.Val386fs
2:25246742:A:AC CGGGAAC	NA	frameshift	c.1149_1156dupGT TCCCGG	p.Val386fs
2:25246742:AC:A	NA	frameshift	c.1156delG	p.Val386fs
2:25246744:C:CG	NA	frameshift	c.1154dupC	p.Val386fs
2:25246744:C:CG GGAACA	NA	frameshift	c.1148_1154dupTG TTCCC	p.Cys387fs
2:25246744:CG:C	rs779418049	frameshift	c.1154delC	p.Pro385fs
2:25246744:CGG: C	NA	frameshift	c.1153_1154delCC	p.Pro385fs
2:25246744:CGG GA:C	NA	frameshift	c.1151_1154delTC CC	p.Phe384fs
2:25246744:CGG GAACAGCTTCCCC GCGCGGCTGCTG: C	NA	inframe_indel	c.1128_1154delCA GCAGCCGCGCGGG GAAGCTGTTCCC	p.Ser377_Pro385 del
2:25246745:GGG AACAGCTTCCCCG C:G	NA	frameshift	c.1138_1153delGCG GGGGAAGCTGTTCC	p.Ala380fs
2:25246747:G:GA		frameshift	c.1151dupT	p.Val386fs
2:25246747:G:GT	rs1196506372	frameshift	c.1151_1152insA	p.Phe384fs
2:25246747:GA:G	NA	frameshift	c.1151delT	p.Phe384fs
2:25246747:GAA CAGCTTCCCCGCG CGGCTGCTGGCC: G	NA	inframe_indel	c.1125_1151delGG CCAGCAGCCGCGC GGGGAAGCTGTT	p.Ala376_Phe384 del
2:25246750:C:CA A	NA	frameshift	c.1148_1149insTT	p.Phe384fs
2:25246750:CA:C	NA	frameshift	c.1148delT	p.Leu383fs
2:25246751:A:AA G	NA	frameshift	c.1147_1148insCT	p.Leu383fs

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2:25246751:AGCT TC:A	NA	frameshift	c.1143_1147delGA AGC	p.Lys382fs
2:25246751:AGCT TCCCC:A	NA	frameshift	c.1140_1147delGG GGAAGC	p.Gly381fs
2:25246753:CT:C	NA	frameshift	c.1145delA	p.Lys382fs
2:25246755:T:A	NA	stop_gained	c.1144A>T	p.Lys382*
2:25246755:TC:T	rs745939351	frameshift	c.1143delG	p.Lys382fs
2:25246758:CCG: C	NA	frameshift	c.1139_1140delCG	p.Ala380fs
2:25246758:CCGC GCGGCTGCTGG:C	NA	frameshift	c.1127_1140delCC AGCAGCCGCGCG	p.Ala376fs
2:25246759:CG:C	NA	frameshift	c.1139delC	p.Ala380fs
2:25246760:G:T	NA	missense	c.1139C>A	p.Ala380Glu
2:25246761:C:CA	NA	frameshift	c.1137_1138insT	p.Ala380fs
2:25246761:C:G	NA	missense	c.1138G>C	p.Ala380Pro
2:25246761:CGC GGCTGCTGGCCAC CT:C	NA	splice_acceptor	c.1123- 2_1137delAGGTGG CCAGCAGCCGC	p.Val375_Ala380 del
2:25246762:G:GC	NA	frameshift	c.1136dupG	p.Ala380fs
2:25246762:GCG GCTGCT:G	NA	frameshift	c.1129_1136delAG CAGCCG	p.Ser377fs
2:25246763:C:A	NA	missense	c.1136G>T	p.Arg379Leu
2:25246763:C:CG	NA	frameshift	c.1135dupC	p.Arg379fs
2:25246763:C:G	NA	missense	c.1136G>C	p.Arg379Pro
2:25246763:C:T	rs1036696061	missense	c.1136G>A	p.Arg379His
2:25246763:CG:C	NA	frameshift	c.1135delC	p.Arg379fs
2:25246764:G:A	rs754459010	missense	c.1135C>T	p.Arg379Cys
2:25246764:G:C	NA	missense	c.1135C>G	p.Arg379Gly
2:25246764:G:T	NA	missense	c.1135C>A	p.Arg379Ser
2:25246765:G:T	NA	missense	c.1134C>A	p.Ser378Arg
2:25246765:GCTG CTGGCCAC:G	NA	frameshift	c.1123_1133delGT GGCCAGCAG	p.Val375fs
2:25246766:CT:C	NA	frameshift	c.1132delA	p.Ser378fs

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2:25246766:CTGC T:C	NA	frameshift	c.1129_1132delAG CA	p.Ser377fs
2:25246767:TG:T	NA	frameshift	c.1131delC	p.Ser377fs
2:25246768:GC:G	NA	frameshift	c.1130delG	p.Ser377fs
2:25246770:TG:T	NA	frameshift	c.1128delC	p.Ser377fs
2:25246770:TGGC CACCTGGA:T	NA	splice_acceptor	c.1123- 5_1128delTCCAGG TGGCC	p.Val375_Ser377 del
2:25246772:G:A	NA	missense	c.1127C>T	p.Ala376Val
2:25246772:G:T	NA	missense	c.1127C>A	p.Ala376Asp
2:25246772:GC:G	rs1383888539	frameshift	c.1126delG	p.Ala376fs
2:25246773:C:G	NA	missense	c.1126G>C	p.Ala376Pro
2:25246773:C:T	rs1446942650	missense	c.1126G>A	p.Ala376Thr
2:25246773:CCA: C	NA	frameshift	c.1124_1125delTG	p.Val375fs
2:25246773:CCAC CTGGAGG:C	NA	splice_acceptor	c.1123- 7_1125delCCTCCA GGTG	p.Val375_Ala376 del
2:25246773:CCAC CTGGAGGGTGA:C	NA	splice_acceptor	c.1123- 11_1125delTCACCC TCCAGGTG	p.Val375_Ala376 del
2:25246775:A:C	NA	missense	c.1124T>G	p.Val375Gly
2:25246775:AC:A	NA	frameshift	c.1123delG	p.Val375fs
2:25246776:CCTG GAGGGTGACA:C	NA	splice_acceptor	c.1123-13_1123- 1delTGTACCCCTCC AG	
2:25246777:C:A	rs781118037	splice_acceptor	c.1123-1G>T	
2:25246777:C:G	NA	splice_acceptor	c.1123-1G>C	
2:25246777:C:T	rs781118037	splice_acceptor	c.1123-1G>A	
2:25246777:CT:C	NA	splice_acceptor	c.1123-2delA	

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2:25246777:CTGG AGGGTGA:C	NA	splice_acceptor	c.1123-11_1123- 2delTCACCTCCA	
2:25246777:CTGG AGGGTGACA:C	NA	splice_acceptor	c.1123-13_1123- 2delTGTCACCTCC A	
2:25246778:T:A	NA	splice_acceptor	c.1123-2A>T	
2:25246778:T:C	rs752605931	splice_acceptor	c.1123-2A>G	
2:25246778:T:G		splice_acceptor	c.1123-2A>C	
2:25247048:CA:C	NA	splice_donor	c.1122+2delT	
2:25247049:A:C	NA	splice_donor	c.1122+2T>G	
2:25247049:A:G	NA	splice_donor	c.1122+2T>C	
2:25247049:A:T	NA	splice_donor	c.1122+2T>A	
2:25247049:AC:A	NA	splice_donor	c.1122+1delG	
2:25247050:C:G	NA	splice_donor	c.1122+1G>C	
2:25247050:C:T	rs747220514	splice_donor	c.1122+1G>A	
2:25247051:CT:C	NA	frameshift	c.1121delA	p.Gln374fs
2:25247052:T:A	NA	missense	c.1121A>T	p.Gln374Leu
2:25247052:TG:T	NA	frameshift	c.1120delC	p.Gln374fs
2:25247053:G:A	rs369109129	stop_gained	c.1120C>T	p.Gln374*
2:25247054:C:CT	NA	frameshift	c.1118_1119insA	p.Gln374fs
2:25247055:A:C	NA	missense	c.1118T>G	p.Leu373Arg
2:25247055:A:G	NA	missense	c.1118T>C	p.Leu373Pro
2:25247055:A:T	rs748329208	missense	c.1118T>A	p.Leu373Gln
2:25247055:AG:A	NA	frameshift	c.1117delC	p.Leu373fs
2:25247055:AGG ACCTCGTAGAT:A	NA	frameshift	c.1105_1117delAT CTACGAGGTCC	p.Ile369fs
2:25247056:G:C	NA	missense	c.1117C>G	p.Leu373Val
2:25247057:G:GA	NA	frameshift	c.1115dupT	p.Leu373fs
2:25247058:A:T	rs773722655	missense	c.1115T>A	p.Val372Asp
2:25247058:AC:A	NA	frameshift	c.1114delG	p.Val372fs

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2:25247058:ACCT :A	NA	inframe_indel	c.1112_1114delAG G	p.Glu371del
2:25247059:C:G	NA	missense	c.1114G>C	p.Val372Leu
2:25247059:C:T	rs371677904	missense	c.1114G>A	p.Val372Ile
2:25247060:CT:C	NA	frameshift	c.1112delA	p.Glu371fs
2:25247061:TC:T	NA	frameshift	c.1111delG	p.Glu371fs
2:25247061:TCGT A:T	NA	frameshift	c.1108_1111delTA CG	p.Tyr370fs
2:25247062:C:A	rs1290621612	stop_gained	c.1111G>T	p.Glu371*
2:25247062:C:T	rs1290621612	missense	c.1111G>A	p.Glu371Lys
2:25247063:G:C	NA	stop_gained	c.1110C>G	p.Tyr370*
2:25247063:G:T	NA	stop_gained	c.1110C>A	p.Tyr370*
2:25247064:T:G	NA	missense	c.1109A>C	p.Tyr370Ser
2:25247064:T:TA	NA	frameshift	c.1108dupT	p.Tyr370fs
2:25247065:AG:A	NA	frameshift	c.1107delC	p.Tyr370fs
2:25247065:AGAT GG:A	NA	frameshift	c.1103_1107delCC ATC	p.Ala368fs
2:25247066:G:GA TGGCTTTGCGGTA C	NA	inframe_indel	c.1092_1106dupGT ACCGCAAAGCCAT	p.Met364_Ala368 dup
2:25247066:GAT GGCTTTGCGGTAC :G	NA	inframe_indel	c.1092_1106delGT ACCGCAAAGCCAT	p.Met364_Ala368 del
2:25247067:A:C	NA	missense	c.1106T>G	p.Ile369Ser
2:25247067:A:T	rs773941479	missense	c.1106T>A	p.Ile369Asn
2:25247068:T:A	NA	missense	c.1105A>T	p.Ile369Phe
2:25247068:TG:T	NA	frameshift	c.1104delC	p.Ile369fs
2:25247070:G:A	rs759087082	missense	c.1103C>T	p.Ala368Val
2:25247070:G:GC	NA	frameshift	c.1102dupG	p.Ala368fs
2:25247070:G:T	NA	missense	c.1103C>A	p.Ala368Asp
2:25247071:C:G	NA	missense	c.1102G>C	p.Ala368Pro
2:25247071:C:T	rs1347653303	missense	c.1102G>A	p.Ala368Thr
2:25247071:CT:C	NA	frameshift	c.1101delA	p.Ala368fs

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2:25247072:T:TTT GCGGTACATGGG CTGCTTG	NA	inframe_indel	c.1080_1100dupCA AGCAGCCCATGTAC CGCAA	p.Asn360_Arg366 dup
2:25247072:TTTG CGGTACATGG:T	NA	frameshift	c.1088_1100delCC ATGTACCGCAA	p.Pro363fs
2:25247074:TG:T	NA	frameshift	c.1098delC	p.Ala368fs
2:25247076:C:A	rs767236033	missense	c.1097G>T	p.Arg366Leu
2:25247076:C:CG GTA	NA	frameshift	c.1093_1096dupTA CC	p.Arg366fs
2:25247076:C:G	NA	missense	c.1097G>C	p.Arg366Pro
2:25247076:C:T	rs767236033	missense	c.1097G>A	p.Arg366His
2:25247076:CG:C	NA	frameshift	c.1096delC	p.Arg366fs
2:25247076:CGGT A:C	NA	frameshift	c.1093_1096delTA CC	p.Tyr365fs
2:25247077:G:A	NA	missense	c.1096C>T	p.Arg366Cys
2:25247077:G:C	NA	missense	c.1096C>G	p.Arg366Gly
2:25247077:G:T	NA	missense	c.1096C>A	p.Arg366Ser
2:25247078:G:C	NA	stop_gained	c.1095C>G	p.Tyr365*
2:25247078:G:T	NA	stop_gained	c.1095C>A	p.Tyr365*
2:25247079:T:C	rs144062658	missense	c.1094A>G	p.Tyr365Cys
2:25247079:T:G	NA	missense	c.1094A>C	p.Tyr365Ser
2:25247079:T:TA C	NA	frameshift	c.1092_1093dupGT	p.Tyr365fs
2:25247079:TA:T	NA	frameshift	c.1093delT	p.Tyr365fs
2:25247080:A:G	rs1315257786	missense	c.1093T>C	p.Tyr365His
2:25247080:AC:A	NA	frameshift	c.1092delG	p.Met364fs
2:25247080:ACAT GG:A	NA	frameshift	c.1088_1092delCC ATG	p.Pro363fs
2:25247083:TG:T	NA	frameshift	c.1089delC	p.Met364fs
2:25247085:G:GG C	NA	frameshift	c.1086_1087dupGC	p.Pro363fs
2:25247088:T:G	NA	missense	c.1085A>C	p.Gln362Pro
2:25247088:TG:T	rs749080312	frameshift	c.1084delC	p.Gln362fs
2:25247089:G:A	rs1490273086	stop_gained	c.1084C>T	p.Gln362*

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2:25247089:G:GC	NA	frameshift	c.1083dupG	p.Gln362fs
2:25247089:GC:G	NA	frameshift	c.1083delG	p.Lys361fs
2:25247090:CT:C	NA	frameshift	c.1082delA	p.Lys361fs
2:25247090:CTTG :C	NA	inframe_indel	c.1080_1082delCA A	p.Asn360del
2:25247090:CTTG TTGT:C	NA	frameshift	c.1076_1082delAC AACAA	p.Tyr359fs
2:25247092:T:A	NA	stop_gained	c.1081A>T	p.Lys361*
2:25247092:T:C	NA	missense	c.1081A>G	p.Lys361Glu
2:25247093:G:GT	NA	frameshift	c.1079dupA	p.Asn360fs
2:25247093:G:GT T	NA	frameshift	c.1078_1079dupAA	p.Asn360fs
2:25247094:T:TT GTA	NA	frameshift	c.1075_1078dupTA CA	p.Asn360fs
2:25247095:TG:T	NA	frameshift	c.1077delC	p.Tyr359fs
2:25247096:G:C	NA	stop_gained	c.1077C>G	p.Tyr359*
2:25247096:G:GT	NA	frameshift	c.1076dupA	p.Tyr359fs
2:25247096:G:T	NA	stop_gained	c.1077C>A	p.Tyr359*
2:25247098:A:T	NA	missense	c.1075T>A	p.Tyr359Asn
2:25247098:AC:A	NA	frameshift	c.1074delG	p.Tyr359fs
2:25247099:CG:C	NA	frameshift	c.1073delC	p.Thr358fs
2:25247100:G:A	rs1191462913	missense	c.1073C>T	p.Thr358Met
2:25247100:G:T	NA	missense	c.1073C>A	p.Thr358Lys
2:25247100:GT:G	NA	frameshift	c.1072delA	p.Thr358fs
2:25247103:GC:G	NA	frameshift	c.1069delG	p.Ala357fs
2:25247104:C:T	NA	missense	c.1069G>A	p.Ala357Thr
2:25247105:CT:C	NA	frameshift	c.1067delA	p.Gln356fs
2:25247106:TG:T	NA	frameshift	c.1066delC	p.Gln356fs
2:25247106:TGGT GGAACGCAC:T	NA	inframe_indel	c.1055_1066delGT GCGTTCCACC	p.Ser352_Gln356 delinsLys
2:25247107:G:A	rs764062059	stop_gained	c.1066C>T	p.Gln356*
2:25247107:GGT GGAACGCA:G	NA	frameshift	c.1056_1065delTG CGTTCAC	p.Ala353fs

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2:25247109:T:TG	NA	frameshift	c.1063dupC	p.His355fs
2:25247109:T:TG G	NA	frameshift	c.1062_1063dupCC	p.His355fs
2:25247109:TG:T	rs1274074419	frameshift	c.1063delC	p.His355fs
2:25247109:TGG AACGCACTGCA:T	NA	frameshift	c.1051_1063delTG CAGTGCGTTCC	p.Cys351fs
2:25247111:G:GA ACGCACTAA	NA	frameshift	c.1061_1062insTTA GTGCGTT	p.His355fs
2:25247111:GA:G	NA	frameshift	c.1061delT	p.Phe354fs
2:25247111:GAA CGCACTGCAA:G	NA	inframe_indel	c.1050_1061delTT GCAGTGCGTT	p.Cys351_Phe354 del
2:25247112:A:C	NA	missense	c.1061T>G	p.Phe354Cys
2:25247112:A:G	NA	missense	c.1061T>C	p.Phe354Ser
2:25247112:AAC GC:A	NA	frameshift	c.1057_1060delGC GT	p.Ala353fs
2:25247113:A:AC G	NA	frameshift	c.1058_1059dupCG	p.Phe354fs
2:25247113:A:C	rs1161125109	missense	c.1060T>G	p.Phe354Val
2:25247113:AC:A	NA	frameshift	c.1059delG	p.Phe354fs
2:25247114:CG:C	NA	frameshift	c.1058delC	p.Ala353fs
2:25247115:G:A	rs1326972729	missense	c.1058C>T	p.Ala353Val
2:25247115:G:GC A	NA	frameshift	c.1056_1057dupTG	p.Ala353fs
2:25247115:GC:G	NA	frameshift	c.1057delG	p.Ala353fs
2:25247116:CA:C	NA	frameshift	c.1056delT	p.Ser352fs
2:25247117:AC:A	NA	frameshift	c.1055delG	p.Ser352fs
2:25247117:ACT: A	NA	frameshift	c.1054_1055delAG	p.Ser352fs
2:25247118:C:T	rs139053291	missense	c.1055G>A	p.Ser352Asn
2:25247118:CT:C	NA	frameshift	c.1054delA	p.Ser352fs
2:25247119:T:C	NA	missense	c.1054A>G	p.Ser352Gly
2:25247119:T:TG CAAAACGAGCTCA GCGGCATCA	NA	frameshift	c.1031_1053dupTG ATGCCGCTGAGCTC GTTTTGC	p.Ser352fs

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2:25247119:TG:T	rs761025051	frameshift	c.1053delC	p.Cys351fs
2:25247120:G:T	NA	stop_gained	c.1053C>A	p.Cys351*
2:25247120:GC:G	NA	frameshift	c.1052delG	p.Cys351fs
2:25247121:C:CA	NA	frameshift	c.1051dupT	p.Cys351fs
2:25247121:CA:C		frameshift	c.1051delT	p.Cys351fs
2:25247121:CAA: C	NA	frameshift	c.1050_1051delTT	p.Phe350fs
2:25247122:AAAA C:A	NA	frameshift	c.1047_1050delGT TT	p.Phe350fs
2:25247124:A:G	NA	missense	c.1049T>C	p.Phe350Ser
2:25247125:AC:A	NA	frameshift	c.1047delG	p.Cys351fs
2:25247127:G:A	NA	missense	c.1046C>T	p.Ser349Leu
2:25247127:G:GA	NA	frameshift	c.1045dupT	p.Ser349fs
2:25247127:G:T	rs1297885007	stop_gained	c.1046C>A	p.Ser349*
2:25247129:GC:G	NA	frameshift	c.1043delG	p.Ser348fs
2:25247130:C:T	NA	missense	c.1043G>A	p.Ser348Asn
2:25247130:CTCA :C	NA	inframe_indel	c.1040_1042delTG A	p.Leu347_Ser348 delinsArg
2:25247130:CTCA GCGGCA:C	NA	inframe_indel	c.1034_1042delTG CCGCTGA	p.Met345_Leu34 7del
2:25247131:T:A	NA	missense	c.1042A>T	p.Ser348Cys
2:25247131:T:TC AGC	NA	frameshift	c.1038_1041dupGC TG	p.Ser348fs
2:25247132:C:CA	NA	frameshift	c.1040dupT	p.Ser348fs
2:25247133:A:C	NA	missense	c.1040T>G	p.Leu347Arg
2:25247133:A:G	rs1474463141	missense	c.1040T>C	p.Leu347Pro
2:25247133:A:T	NA	missense	c.1040T>A	p.Leu347Gln
2:25247133:AG:A	NA	frameshift	c.1039delC	p.Leu347fs
2:25247133:AGC G:A	NA	inframe_indel	c.1037_1039delCG C	p.Pro346del
2:25247135:CG:C	NA	frameshift	c.1037delC	p.Pro346fs
2:25247136:G:A	rs1308281604	missense	c.1037C>T	p.Pro346Leu

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2:25247136:G:GG CATC	NA	frameshift	c.1032_1036dupG ATGC	p.Pro346fs
2:25247137:G:GC	NA	frameshift	c.1035dupG	p.Pro346fs
2:25247137:G:T	NA	missense	c.1036C>A	p.Pro346Thr
2:25247138:CA:C	NA	frameshift	c.1034delT	p.Met345fs
2:25247139:A:AC	NA	frameshift	c.1033_1034insG	p.Met345fs
2:25247139:AT:A	NA	frameshift	c.1033delA	p.Met345fs
2:25247139:ATCA GCTTCTCAAC:A	NA	frameshift	c.1021_1033delGT TGAGAAGCTGA	p.Val341fs
2:25247141:C:CA T	NA	frameshift	c.1031_1032insAT	p.Met345fs
2:25247141:CA:C	NA	frameshift	c.1031delT	p.Leu344fs
2:25247141:CAGC TTCTCAACACACA: C	NA	frameshift	c.1016_1031delTG TGTGTTGAGAAGCT	p.Val339fs
2:25247142:A:C	rs749817324	missense	c.1031T>G	p.Leu344Arg
2:25247142:A:G	rs749817324	missense	c.1031T>C	p.Leu344Pro
2:25247142:A:T	rs749817324	missense	c.1031T>A	p.Leu344Gln
2:25247142:AG:A	NA	frameshift	c.1030delC	p.Leu344fs
2:25247143:G:T	NA	missense	c.1030C>A	p.Leu344Met
2:25247144:CT:C	NA	frameshift	c.1028delA	p.Lys343fs
2:25247145:T:C	NA	missense	c.1028A>G	p.Lys343Arg
2:25247146:T:A	NA	stop_gained	c.1027A>T	p.Lys343*
2:25247146:T:TG TCCCCCAGGTGT GTGTTGAGAAG	NA	frameshift	c.1026_1027insCTT CTCAACACACACCT GGGGGGAC	p.Lys343fs
2:25247147:CT:C	NA	frameshift	c.1025delA	p.Glu342fs
2:25247148:TC:T	NA	frameshift	c.1024delG	p.Glu342fs
2:25247149:C:A	NA	stop_gained	c.1024G>T	p.Glu342*
2:25247149:CAAC A:C	NA	frameshift	c.1020_1023delTG TT	p.Cys340fs
2:25247150:A:AA C	rs768966400	frameshift	c.1021_1022dupGT	p.Glu342fs

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2:25247150:AAC: A	rs1201885886	frameshift	c.1021_1022delGT	p.Val341fs
2:25247150:AACA C:A	NA	frameshift	c.1019_1022delGT GT	p.Cys340fs
2:25247151:A:AC ACACAC	NA	frameshift	c.1015_1021dupGT GTGTG	p.Val341fs
2:25247151:AC:A	NA	frameshift	c.1021delG	p.Val341fs
2:25247151:ACAC ACACCT:A	NA	frameshift	c.1015- 2_1021delAGGTGT GTG	p.Val339fs
2:25247152:CACA CACCTGGGGG:C	NA	splice_acceptor	c.1015- 7_1020delCCCCCA GGTGTGT	p.Val339_Val341del
2:25247153:A:T	NA	stop_gained	c.1020T>A	p.Cys340*
2:25247153:AC:A	NA	frameshift	c.1019delG	p.Cys340fs
2:25247153:ACAC AC:A	NA	frameshift	c.1015_1019delGT GTG	p.Val339fs
2:25247156:C:CA	NA	frameshift	c.1016dupT	p.Cys340fs
2:25247156:C:CA CCT	NA	frameshift	c.1015- 2_1016dupAGGT	p.Cys340fs
2:25247156:CA:C	NA	frameshift	c.1016delT	p.Val339fs
2:25247156:CACC TG:C	NA	frameshift	c.1015- 3_1016delCAGGT	p.Val341fs
2:25247156:CACC TGGGGGGG:C	NA	frameshift	c.1015- 8_1016delCCCCC AGGT	p.Val341fs
2:25247157:A:AC	rs773201456	frameshift	c.1015dupG	p.Val339fs
2:25247157:A:C	rs1221845015	missense	c.1016T>G	p.Val339Gly
2:25247157:AC:A	rs34203089	frameshift	c.1015delG	p.Val339fs
2:25247159:C:A	rs1458692279	splice_acceptor	c.1015-1G>T	
2:25247159:C:G	NA	splice_acceptor	c.1015-1G>C	

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2:25247159:C:T	NA	splice_acceptor	c.1015-1G>A	
2:25247159:CT:C	rs762753119	splice_acceptor	c.1015-2delA	
2:25247160:T:A	rs920946076	splice_acceptor	c.1015-2A>T	
2:25247160:T:C	rs920946076	splice_acceptor	c.1015-2A>G	
2:25247160:T:G	NA	splice_acceptor	c.1015-2A>C	
2:25247588:T:TA	NA	splice_donor	c.1014+2dupT	
2:25247588:TACC:T	NA	frameshift	c.1014_1014+2delGGT	p.Val339fs
2:25247589:A:C	rs1258308759	splice_donor	c.1014+2T>G	
2:25247589:A:G	NA	splice_donor	c.1014+2T>C	
2:25247589:A:T	NA	splice_donor	c.1014+2T>A	
2:25247589:AC:A	NA	splice_donor	c.1014+1delG	
2:25247589:ACCACTG:A	NA	frameshift	c.1010_1014+1delCAGTGG	p.Ser337fs
2:25247590:C:A	NA	splice_donor	c.1014+1G>T	
2:25247590:C:G	NA	splice_donor	c.1014+1G>C	
2:25247590:C:T	NA	splice_donor	c.1014+1G>A	
2:25247590:CCA:C	NA	frameshift	c.1013_1014delTG	p.Val338fs
2:25247591:CA:C	NA	frameshift	c.1013delT	p.Val338fs
2:25247591:CACTGAGA:C	NA	frameshift	c.1007_1013delTCTCAGT	p.Phe336fs
2:25247593:C:CTGAGAATTTGCCGTCTCCGAACCACA	NA	frameshift	c.986_1011dupTGTTGGTTCGGAGACGGCAAATTCTCA	p.Val338fs
2:25247593:C:T	NA	missense	c.1012G>A	p.Val338Met
2:25247594:T:TGA	NA	frameshift	c.1009_1010dupTC	p.Val338fs
2:25247594:TG:T	rs1212817360	frameshift	c.1010delC	p.Ser337fs

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2:25247595:G:A	rs773208295	missense	c.1010C>T	p.Ser337Leu
2:25247595:G:C	rs773208295	stop_gained	c.1010C>G	p.Ser337*
2:25247595:G:T	rs773208295	stop_gained	c.1010C>A	p.Ser337*
2:25247595:GAG A:G	NA	inframe_indel	c.1007_1009delTCT	p.Phe336del
2:25247596:A:G	NA	missense	c.1009T>C	p.Ser337Pro
2:25247597:G:GA T	NA	frameshift	c.1007_1008insAT	p.Phe336fs
2:25247597:GA:G	NA	frameshift	c.1007delT	p.Phe336fs
2:25247597:GAA: G	NA	frameshift	c.1006_1007delTT	p.Phe336fs
2:25247597:GAAT T:G	NA	frameshift	c.1004_1007delAA TT	p.Lys335fs
2:25247598:A:AA TT	NA	stop_gained	c.1004_1006dupAA T	p.Lys335_Phe336i nsTer
2:25247598:AATT TGCCGTCTCC:A	NA	frameshift	c.994_1006delGGA GACGGCAAAT	p.Gly332fs
2:25247599:AT:A	NA	frameshift	c.1005delA	p.Lys335fs
2:25247600:TTTG CCGTCTCCGAACC ACATGAC:T	NA	frameshift	c.982_1004delGTC ATGTGGTTCGGAGA CGGCAA	p.Val328fs
2:25247602:T:A	NA	stop_gained	c.1003A>T	p.Lys335*
2:25247603:GC:G	NA	frameshift	c.1001delG	p.Gly334fs
2:25247605:C:CT	NA	frameshift	c.999_1000insA	p.Gly334fs
2:25247605:C:T	rs1024296111	missense	c.1000G>A	p.Gly334Ser
2:25247605:CG:C	NA	frameshift	c.999delC	p.Asp333fs
2:25247605:CGTC T:C	NA	frameshift	c.996_999delAGAC	p.Asp333fs
2:25247606:GTC: G	NA	frameshift	c.997_998delGA	p.Asp333fs
2:25247607:T:A	NA	missense	c.998A>T	p.Asp333Val
2:25247608:C:CT CCG	NA	frameshift	c.993_996dupCGG A	p.Asp333fs

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2:25247608:CTCC GA:C	NA	frameshift	c.992_996delTCGG A	p.Phe331fs
2:25247609:TCCG AACCACATGACCC AGCGGG:T	NA	frameshift	c.974_995delCCCG CTGGGTCATGTGGT TCGG	p.Thr325fs
2:25247610:C:A	NA	missense	c.995G>T	p.Gly332Val
2:25247610:C:T	rs751360082	missense	c.995G>A	p.Gly332Glu
2:25247611:C:A	rs760854242	stop_gained	c.994G>T	p.Gly332*
2:25247611:C:T	rs760854242	missense	c.994G>A	p.Gly332Arg
2:25247611:CG:C	NA	frameshift	c.993delC	p.Phe331fs
2:25247612:GA:G	NA	frameshift	c.992delT	p.Phe331fs
2:25247613:AACC :A	rs755832478	inframe_indel	c.989_991delGGT	p.Trp330del
2:25247614:A:C	rs753896945	missense	c.991T>G	p.Phe331Val
2:25247614:A:T	NA	missense	c.991T>A	p.Phe331Ile
2:25247614:AC:A	rs777388989	frameshift	c.990delG	p.Trp330fs
2:25247615:C:A	NA	missense	c.990G>T	p.Trp330Cys
2:25247615:C:T	rs757340349	stop_gained	c.990G>A	p.Trp330*
2:25247615:CCAC ATG:C	NA	inframe_indel	c.984_989delCATG TG	p.Met329_Trp330 del
2:25247615:CCAC ATGACCCA:C	NA	frameshift	c.979_989delTGGG TCATGTG	p.Trp327fs
2:25247616:C:A	NA	missense	c.989G>T	p.Trp330Leu
2:25247616:C:T	rs1479473205	stop_gained	c.989G>A	p.Trp330*
2:25247616:CACA TG:C	NA	frameshift	c.984_988delCATG T	p.Met329fs
2:25247617:ACAT GACCCAG:A	NA	frameshift	c.978_987delCTGG GTCATG	p.Trp327fs
2:25247618:C:CA	NA	frameshift	c.986dupT	p.Met329fs
2:25247618:C:T	NA	missense	c.987G>A	p.Met329Ile
2:25247619:A:G	NA	missense	c.986T>C	p.Met329Thr
2:25247619:AT:A	NA	frameshift	c.985delA	p.Met329fs
2:25247621:GA:G	NA	frameshift	c.983delT	p.Val328fs
2:25247622:A:AC	NA	frameshift	c.982dupG	p.Val328fs

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2:25247622:A:C	NA	missense	c.983T>G	p.Val328Gly
2:25247622:A:G	NA	missense	c.983T>C	p.Val328Ala
2:25247622:A:T	NA	missense	c.983T>A	p.Val328Asp
2:25247622:ACCC AGCGGGTG:A	NA	frameshift	c.972_982delCACC CGCTGGG	p.Thr325fs
2:25247623:C:A	NA	missense	c.982G>T	p.Val328Phe
2:25247624:C:T	NA	stop_gained	c.981G>A	p.Trp327*
2:25247625:C:A	NA	missense	c.980G>T	p.Trp327Leu
2:25247625:C:CA	NA	frameshift	c.979dupT	p.Trp327fs
2:25247625:C:T	rs750966422	stop_gained	c.980G>A	p.Trp327*
2:25247625:CAGC GGGTGCCTTCA:C	NA	frameshift	c.966_979delTGAA GGCACCCGCT	p.Glu323fs
2:25247626:AG:A	NA	frameshift	c.978delC	p.Trp327fs
2:25247627:GC:G	NA	frameshift	c.977delG	p.Arg326fs
2:25247627:GCG GGTGC:G	NA	frameshift	c.971_977delGCAC CCG	p.Gly324fs
2:25247628:C:A	rs758881009	missense	c.977G>T	p.Arg326Leu
2:25247628:C:CA GCTGAAGGCACCC GCT	NA	frameshift	c.976_977insAGCG GGTGCCTTCAGCT	p.Arg326fs
2:25247628:C:CG	NA	frameshift	c.976dupC	p.Arg326fs
2:25247628:C:G	NA	missense	c.977G>C	p.Arg326Pro
2:25247628:C:T	rs758881009	missense	c.977G>A	p.Arg326His
2:25247628:CG:C	NA	frameshift	c.976delC	p.Arg326fs
2:25247629:G:A	rs747448117	missense	c.976C>T	p.Arg326Cys
2:25247629:G:C	NA	missense	c.976C>G	p.Arg326Gly
2:25247629:G:T	rs747448117	missense	c.976C>A	p.Arg326Ser
2:25247631:GT:G	NA	frameshift	c.973delA	p.Thr325fs
2:25247631:GTGC CTTCAGC:G	NA	frameshift	c.964_973delGCTG AAGGCA	p.Ala322fs
2:25247632:T:TG	NA	frameshift	c.972dupC	p.Thr325fs
2:25247632:TG:T	NA	frameshift	c.972delC	p.Thr325fs
2:25247633:G:GC	NA	frameshift	c.971dupG	p.Thr325fs

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2:25247633:GC:G	NA	frameshift	c.971delG	p.Gly324fs
2:25247634:C:A	NA	missense	c.971G>T	p.Gly324Val
2:25247637:TCAG CTGCTCGGCTCCG GCCCCG:T	NA	inframe_indel	c.947_967delCGGG CCGGAGCCGAGCA GCTG	p.Thr316_Glu323 delinsLys
2:25247638:C:A	NA	stop_gained	c.967G>T	p.Glu323*
2:25247638:CA:C	NA	frameshift	c.966delT	p.Glu323fs
2:25247639:AG:A	NA	frameshift	c.965delC	p.Ala322fs
2:25247640:GCTG CTCGGCTC:G	NA	frameshift	c.954_964delGAGC CGAGCAG	p.Ser319fs
2:25247641:C:CT	NA	frameshift	c.963dupA	p.Ala322fs
2:25247641:CT:C	NA	frameshift	c.963delA	p.Ala322fs
2:25247642:TG:T	NA	frameshift	c.962delC	p.Ala321fs
2:25247644:C:CT CGGT	NA	frameshift	c.960_961insACCG A	p.Ala321fs
2:25247646:C:T	NA	missense	c.959G>A	p.Arg320Gln
2:25247646:CG:C	NA	frameshift	c.958delC	p.Arg320fs
2:25247647:G:A	rs778270132	stop_gained	c.958C>T	p.Arg320*
2:25247648:G:T	NA	missense	c.957C>A	p.Ser319Arg
2:25247649:CT:C	NA	frameshift	c.955delA	p.Ser319fs
2:25247650:T:TCC GGC	NA	frameshift	c.950_954dupGCC GG	p.Ser319fs
2:25247650:TC:T	NA	frameshift	c.954delG	p.Ser319fs
2:25247652:C:CG	NA	frameshift	c.952dupC	p.Arg318fs
2:25247652:CG:C	NA	frameshift	c.952delC	p.Arg318fs
2:25247652:CGG: C	NA	frameshift	c.951_952delCC	p.Arg318fs
2:25247654:G:GC	NA	frameshift	c.950dupG	p.Arg318fs
2:25247654:GC:G	NA	frameshift	c.950delG	p.Gly317fs
2:25247654:GCCC GT:G	NA	frameshift	c.946_950delACGG G	p.Thr316fs
2:25247655:CCCG TCAT:C	NA	frameshift	c.943_949delATGA CGG	p.Met315fs
2:25247658:GT:G	NA	frameshift	c.946delA	p.Thr316fs

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2:25247661:AT:A	NA	frameshift	c.943delA	p.Met315fs
2:25247661:ATCC AC:A	NA	frameshift	c.939_943delGTGG A	p.Trp313fs
2:25247662:TC:T	NA	frameshift	c.942delG	p.Trp314fs
2:25247662:TCC:T	NA	frameshift	c.941_942delGG	p.Trp314fs
2:25247662:TCCA CCAA:T	NA	frameshift	c.936_942delTTGG TGG	p.Trp313fs
2:25247662:TCCA CCAAGACACAA:T	NA	frameshift	c.929_942delTTGT GTCTTGGTGG	p.Ile310fs
2:25247663:C:T	rs772972939	stop_gained	c.942G>A	p.Trp314*
2:25247663:CCA: C	NA	frameshift	c.940_941delTG	p.Trp314fs
2:25247663:CCAC CAAGA:C	NA	frameshift	c.934_941delTCTT GGTG	p.Ser312fs
2:25247664:C:T	rs749225216	stop_gained	c.941G>A	p.Trp314*
2:25247664:CA:C	NA	frameshift	c.940delT	p.Trp314fs
2:25247665:A:AC	NA	frameshift	c.939dupG	p.Trp314fs
2:25247665:A:AC C	NA	frameshift	c.938_939dupGG	p.Trp314fs
2:25247665:AC:A	NA	frameshift	c.939delG	p.Trp313fs
2:25247666:C:T	rs770813937	stop_gained	c.939G>A	p.Trp313*
2:25247667:C:CA	NA	frameshift	c.937dupT	p.Trp313fs
2:25247667:C:T	rs1204478457	stop_gained	c.938G>A	p.Trp313*
2:25247667:CA:C	NA	frameshift	c.937delT	p.Trp313fs
2:25247670:G:A	NA	missense	c.935C>T	p.Ser312Phe
2:25247670:G:T	NA	missense	c.935C>A	p.Ser312Tyr
2:25247670:GA:G	NA	frameshift	c.934delT	p.Ser312fs
2:25247670:GAC: G	NA	frameshift	c.933_934delGT	p.Ser312fs
2:25247671:AC:A	NA	frameshift	c.933delG	p.Ser312fs
2:25247672:C:CA	NA	frameshift	c.932dupT	p.Ser312fs
2:25247672:CA:C	NA	frameshift	c.932delT	p.Val311fs
2:25247673:A:C	NA	missense	c.932T>G	p.Val311Gly

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2:25247674:C:A	NA	missense	c.931G>T	p.Val311Leu
2:25247674:C:CA	NA	frameshift	c.930dupT	p.Val311fs
2:25247674:C:CA A	NA	frameshift	c.929_930dupTT	p.Val311fs
2:25247674:CA:C	NA	frameshift	c.930delT	p.Ile310fs
2:25247675:A:C	NA	missense	c.930T>G	p.Ile310Met
2:25247676:A:C	NA	missense	c.929T>G	p.Ile310Ser
2:25247676:A:G	NA	missense	c.929T>C	p.Ile310Thr
2:25247676:A:T	rs587777508	missense	c.929T>A	p.Ile310Asn
2:25247676:AT:A	NA	frameshift	c.928delA	p.Ile310fs
2:25247677:T:A	rs774128516	missense	c.928A>T	p.Ile310Phe
2:25247677:T:G	NA	missense	c.928A>C	p.Ile310Leu
2:25247679:C:G		missense	c.926G>C	p.Arg309Pro
2:25247679:CG:C	NA	frameshift	c.925delC	p.Arg309fs
2:25247680:G:A	NA	missense	c.925C>T	p.Arg309Cys
2:25247680:G:C	NA	missense	c.925C>G	p.Arg309Gly
2:25247680:G:GG C	NA	frameshift	c.923_924dupGC	p.Arg309fs
2:25247681:GC:G	NA	frameshift	c.923delG	p.Gly308fs
2:25247682:C:A	NA	missense	c.923G>T	p.Gly308Val
2:25247682:C:T	NA	missense	c.923G>A	p.Gly308Asp
2:25247683:C:CT	NA	frameshift	c.921dupA	p.Gly308fs
2:25247683:C:G	NA	missense	c.922G>C	p.Gly308Arg
2:25247683:C:T	NA	missense	c.922G>A	p.Gly308Ser
2:25247683:CT:C	NA	frameshift	c.921delA	p.Gly308fs
2:25247684:TG:T	NA	frameshift	c.920delC	p.Pro307fs
2:25247684:TGGC CACCAGGAGAA:T	NA	frameshift	c.907_920delTTCTC CTGGTGGCC	p.Phe303fs
2:25247685:G:A	rs759380437	missense	c.920C>T	p.Pro307Leu
2:25247685:G:C	rs759380437	missense	c.920C>G	p.Pro307Arg
2:25247685:G:T	rs759380437	missense	c.920C>A	p.Pro307Gln
2:25247686:G:A	NA	missense	c.919C>T	p.Pro307Ser
2:25247686:G:C	NA	missense	c.919C>G	p.Pro307Ala

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2:25247686:G:T	NA	missense	c.919C>A	p.Pro307Thr
2:25247686:GC:G	NA	frameshift	c.918delG	p.Trp306fs
2:25247686:GCC: G	NA	frameshift	c.917_918delGG	p.Trp306fs
2:25247687:C:A	rs776841024	missense	c.918G>T	p.Trp306Cys
2:25247687:C:CC A	NA	frameshift	c.916_917dupTG	p.Trp306fs
2:25247687:C:G	NA	missense	c.918G>C	p.Trp306Cys
2:25247687:C:T	rs776841024	stop_gained	c.918G>A	p.Trp306*
2:25247688:C:A	NA	missense	c.917G>T	p.Trp306Leu
2:25247688:C:G	NA	missense	c.917G>C	p.Trp306Ser
2:25247688:C:T	rs1465829182	stop_gained	c.917G>A	p.Trp306*
2:25247688:CA:C	NA	frameshift	c.916delT	p.Trp306fs
2:25247689:A:C	NA	missense	c.916T>G	p.Trp306Gly
2:25247689:AC:A	NA	frameshift	c.915delG	p.Trp305fs
2:25247689:ACCA G:A	NA	frameshift	c.912_915delCTGG	p.Trp305fs
2:25247690:C:A	NA	missense	c.915G>T	p.Trp305Cys
2:25247690:C:T	rs761839006	stop_gained	c.915G>A	p.Trp305*
2:25247691:C:CA GGAGAA	NA	frameshift	c.907_913dupTTCT CCT	p.Trp305fs
2:25247691:C:G	NA	missense	c.914G>C	p.Trp305Ser
2:25247691:C:T	rs765341003	stop_gained	c.914G>A	p.Trp305*
2:25247691:CA:C	NA	frameshift	c.913delT	p.Trp305fs
2:25247691:CAG: C	NA	frameshift	c.912_913delCT	p.Trp305fs
2:25247692:A:G	NA	missense	c.913T>C	p.Trp305Arg
2:25247693:G:GA	NA	frameshift	c.911_912insT	p.Trp305fs
2:25247693:GGA: G	NA	frameshift	c.910_911delTC	p.Ser304fs
2:25247694:G:A	NA	missense	c.911C>T	p.Ser304Phe
2:25247694:GA:G	NA	frameshift	c.910delT	p.Ser304fs
2:25247695:AG:A	NA	frameshift	c.909delC	p.Ser304fs

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2:25247696:G:C	NA	missense	c.909C>G	p.Phe303Leu
2:25247696:GA:G	NA	frameshift	c.908delT	p.Phe303fs
2:25247699:GC:G	NA	frameshift	c.905delG	p.Gly302fs
2:25247700:C:T	NA	missense	c.905G>A	p.Gly302Asp
2:25247701:C:A	NA	missense	c.904G>T	p.Gly302Cys
2:25247701:C:T	rs377670596	missense	c.904G>A	p.Gly302Ser
2:25247703:C:T	NA	missense	c.902G>A	p.Arg301Gln
2:25247703:CG:C	NA	frameshift	c.901delC	p.Arg301fs
2:25247704:G:A	NA	missense	c.901C>T	p.Arg301Trp
2:25247704:GC:G	NA	frameshift	c.900delG	p.Arg301fs
2:25247704:GCA GTTTCCCC:G	NA	frameshift	c.891_900delGGGG AAACTG	p.Trp297fs
2:25247705:CA:C	NA	frameshift	c.899delT	p.Leu300fs
2:25247706:A:T	NA	missense	c.899T>A	p.Leu300Gln
2:25247707:G:T	NA	missense	c.898C>A	p.Leu300Met
2:25247707:GT:G		frameshift	c.897delA	p.Lys299fs
2:25247708:T:A	NA	missense	c.897A>T	p.Lys299Asn
2:25247708:T:G	NA	missense	c.897A>C	p.Lys299Asn
2:25247709:T:G	NA	missense	c.896A>C	p.Lys299Thr
2:25247710:T:A	rs766858016	stop_gained	c.895A>T	p.Lys299*
2:25247710:T:C	NA	missense	c.895A>G	p.Lys299Glu
2:25247710:T:G	rs766858016	missense	c.895A>C	p.Lys299Gln
2:25247710:T:TC	rs1157280960	frameshift	c.894dupG	p.Lys299fs
2:25247710:TC:T	NA	frameshift	c.894delG	p.Lys299fs
2:25247712:C:T	rs1410828051	missense	c.893G>A	p.Gly298Glu
2:25247713:C:A	rs797044904	missense	c.892G>T	p.Gly298Trp
2:25247713:C:G	NA	missense	c.892G>C	p.Gly298Arg
2:25247713:C:T	rs797044904	missense	c.892G>A	p.Gly298Arg
2:25247714:C:A	NA	missense	c.891G>T	p.Trp297Cys
2:25247714:C:G	NA	missense	c.891G>C	p.Trp297Cys
2:25247714:C:T	rs1442214131	stop_gained	c.891G>A	p.Trp297*
2:25247715:C:G	NA	missense	c.890G>C	p.Trp297Ser
2:25247715:C:T	rs944608317	stop_gained	c.890G>A	p.Trp297*

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2:25247716:A:AC	NA	frameshift	c.888dupG	p.Trp297fs
2:25247716:A:C	rs751916447	missense	c.889T>G	p.Trp297Gly
2:25247716:A:G	rs751916447	missense	c.889T>C	p.Trp297Arg
2:25247716:A:T	NA	missense	c.889T>A	p.Trp297Arg
2:25247716:AC:A	NA	frameshift	c.888delG	p.Trp297fs
2:25247716:ACAC CAGCTCC:A	NA	frameshift	c.879_888delGGAG CTGGTG	p.Glu294fs
2:25247716:ACAC CAGCTCCC:A	NA	frameshift	c.878_888delGGGA GCTGGTG	p.Gly293fs
2:25247718:A:AC	NA	frameshift	c.886dupG	p.Val296fs
2:25247718:A:C	rs1402705749	missense	c.887T>G	p.Val296Gly
2:25247718:A:T	rs1402705749	missense	c.887T>A	p.Val296Glu
2:25247718:AC:A	NA	frameshift	c.886delG	p.Val296fs
2:25247719:C:A	NA	missense	c.886G>T	p.Val296Leu
2:25247719:C:G	rs755398725	missense	c.886G>C	p.Val296Leu
2:25247719:C:T	rs755398725	missense	c.886G>A	p.Val296Met
2:25247721:A:AG C	NA	frameshift	c.882_883dupGC	p.Leu295fs
2:25247721:A:C	NA	missense	c.884T>G	p.Leu295Arg
2:25247721:A:G	NA	missense	c.884T>C	p.Leu295Pro
2:25247721:A:T	NA	missense	c.884T>A	p.Leu295Gln
2:25247721:AG:A	NA	frameshift	c.883delC	p.Leu295fs
2:25247722:G:C	rs780953840	missense	c.883C>G	p.Leu295Val
2:25247722:G:GA	NA	frameshift	c.882_883insT	p.Leu295fs
2:25247722:G:T	NA	missense	c.883C>A	p.Leu295Met
2:25247722:GCTC C:G	NA	frameshift	c.879_882delGGAG	p.Glu294fs
2:25247724:T:C	NA	missense	c.881A>G	p.Glu294Gly
2:25247724:TC:T	NA	frameshift	c.880delG	p.Glu294fs
2:25247725:C:A	rs755769867	stop_gained	c.880G>T	p.Glu294*
2:25247725:CCCC AA:C	NA	frameshift	c.875_879delTTGG G	p.Ile292fs

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2:25247726:C:CC CAATG	NA	inframe_indel	c.873_878dupCATT GG	p.Gly293_Glu294i nsIleGly
2:25247727:C:A	NA	missense	c.878G>T	p.Gly293Val
2:25247727:C:T	NA	missense	c.878G>A	p.Gly293Glu
2:25247727:CCAA TGCCAA:C	NA	inframe_indel	c.869_877delTTGG CATTG	p.Phe290_Gly293 delinsTrp
2:25247728:C:A	NA	missense	c.877G>T	p.Gly293Trp
2:25247728:C:CA	NA	frameshift	c.876dupT	p.Gly293fs
2:25247728:C:CA ATGCCAAAGCCCC GGCCGTCCTGGA GCCCCAAGGAGC AGA	NA	frameshift	c.856- 20_876dupTCTGCT CCTTGGGGCTCCAG GACGGCCGGGGCT TTGGCATT	p.Gly293fs
2:25247728:C:G	NA	missense	c.877G>C	p.Gly293Arg
2:25247728:C:T	NA	missense	c.877G>A	p.Gly293Arg
2:25247728:CA:C	rs1271476754	frameshift	c.876delT	p.Ile292fs
2:25247730:A:G	rs777306476	missense	c.875T>C	p.Ile292Thr
2:25247730:AT:A	NA	frameshift	c.874delA	p.Ile292fs
2:25247730:ATG: A	NA	frameshift	c.873_874delCA	p.Ile292fs
2:25247734:CA:C	NA	frameshift	c.870delT	p.Phe290fs
2:25247735:A:AC	NA	frameshift	c.869_870insG	p.Phe290fs
2:25247736:A:AA GCCCCGGCCGTCC TGG	NA	frameshift	c.856- 4_868dupCCAGGA CGGCCGGGGCT	p.Phe290fs
2:25247736:A:C	NA	missense	c.869T>G	p.Phe290Cys
2:25247736:A:G	NA	missense	c.869T>C	p.Phe290Ser
2:25247737:A:C	NA	missense	c.868T>G	p.Phe290Val
2:25247737:A:T	NA	missense	c.868T>A	p.Phe290Ile
2:25247737:AG:A	rs746332586	frameshift	c.867delC	p.Phe290fs
2:25247738:G:GC	NA	frameshift	c.866dupG	p.Phe290fs
2:25247738:GC:G	NA	frameshift	c.866delG	p.Gly289fs
2:25247738:GCC: G	NA	frameshift	c.865_866delGG	p.Gly289fs

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2:25247741:C:CA	NA	frameshift	c.863_864insT	p.Phe290fs
2:25247742:CGG:C	NA	frameshift	c.861_862delCC	p.Arg288fs
2:25247743:GGC CGTCCTGGA:G	NA	splice_acceptor	c.856-5_861delTCCAGGACGGC	p.Asp286_Arg288del
2:25247744:GC:G	NA	frameshift	c.860delG	p.Gly287fs
2:25247746:CG:C	NA	frameshift	c.858delC	p.Asp286fs
2:25247748:TC:T	NA	frameshift	c.856delG	p.Asp286fs
2:25247749:C:CC TGG	NA	splice_acceptor	c.856-4_856-1dupCCAG	
2:25247750:C:A	rs1179790183	splice_acceptor	c.856-1G>T	
2:25247750:C:G	NA	splice_acceptor	c.856-1G>C	
2:25247750:C:T	rs1179790183	splice_acceptor	c.856-1G>A	
2:25247750:CTGG A:C	NA	splice_acceptor	c.856-5_856-2delTCCA	
2:25247751:T:A	NA	splice_acceptor	c.856-2A>T	
2:25247751:T:C	rs1234716262	splice_acceptor	c.856-2A>G	
2:25247751:T:G	rs1234716262	splice_acceptor	c.856-2A>C	
2:25248034:CACC TCGT:C	NA	frameshift	c.851_855+2delACGAGGT	p.Tyr284fs
2:25248035:A:C	NA	splice_donor	c.855+2T>G	
2:25248035:A:G	rs745824601	splice_donor	c.855+2T>C	
2:25248035:AC:A	NA	splice_donor	c.855+1delG	
2:25248036:C:A	rs772041639	splice_donor	c.855+1G>T	
2:25248036:C:G	NA	splice_donor	c.855+1G>C	
2:25248036:C:T	rs772041639	splice_donor	c.855+1G>A	
2:25248037:CT:C	NA	frameshift	c.854delA	p.Glu285fs

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2:25248039:C:A	rs201882909	stop_gained	c.853G>T	p.Glu285*
2:25248039:CG:C	NA	frameshift	c.852delC	p.Tyr284fs
2:25248040:G:C	NA	stop_gained	c.852C>G	p.Tyr284*
2:25248040:G:T	NA	stop_gained	c.852C>A	p.Tyr284*
2:25248040:GT:G	NA	frameshift	c.851delA	p.Tyr284fs
2:25248041:TA:T	NA	frameshift	c.850delT	p.Tyr284fs
2:25248041:TAC: T	NA	frameshift	c.849_850delGT	p.Tyr284fs
2:25248042:A:AC TCTGGCT	NA	frameshift	c.842_849dupAGC CAGAG	p.Tyr284fs
2:25248044:T:TCT GGCTCGTCATCG	NA	frameshift	c.834_847dupCGA TGACGAGCCAG	p.Glu283fs
2:25248044:TC:T	NA	frameshift	c.847delG	p.Glu283fs
2:25248045:C:A	NA	stop_gained	c.847G>T	p.Glu283*
2:25248045:C:CC A	NA	frameshift	c.846_847insTG	p.Glu283fs
2:25248045:C:CT	rs747075883	frameshift	c.846dupA	p.Glu283fs
2:25248049:C:CT CGT	NA	frameshift	c.839_842dupACG A	p.Pro282fs
2:25248050:TC:T	NA	frameshift	c.841delG	p.Glu281fs
2:25248051:C:A	rs773312511	stop_gained	c.841G>T	p.Glu281*
2:25248051:CG:C	NA	frameshift	c.840delC	p.Asp280fs
2:25248052:GT:G	NA	frameshift	c.839delA	p.Asp280fs
2:25248053:TCAT CGCCTGCTTTGGT GGCATTCTTGTC:T	NA	frameshift	c.811_838delGACA AGAATGCCACCAAA GCAGGCGATG	p.Asp271fs
2:25248056:TC:T	NA	frameshift	c.835delG	p.Asp279fs
2:25248062:GCTT TGGTGGCATT:G	NA	frameshift	c.817_829delAATG CCACCAAAG	p.Asn273fs
2:25248063:C:CT	NA	frameshift	c.828dupA	p.Ala277fs
2:25248063:CT:C	NA	frameshift	c.828delA	p.Ala277fs
2:25248063:CTTT GG:C	NA	frameshift	c.824_828delCCAA A	p.Thr275fs
2:25248066:T:A	NA	stop_gained	c.826A>T	p.Lys276*

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2:25248066:TG:T	NA	frameshift	c.825delC	p.Ala277fs
2:25248068:G:GT GGCATTG	NA	frameshift	c.816_823dupGAA TGCCA	p.Thr275fs
2:25248068:GT:G	NA	frameshift	c.823delA	p.Thr275fs
2:25248069:T:TG GCATTCTGTCCC CAGCATCGGACC	NA	frameshift	c.797_822dupGGT CCGATGCTGGGA CAAGAATGCC	p.Thr275fs
2:25248069:TG:T	NA	frameshift	c.822delC	p.Thr275fs
2:25248071:GC:G	NA	frameshift	c.820delG	p.Ala274fs
2:25248072:CA:C	NA	frameshift	c.819delT	p.Asn273fs
2:25248073:AT:A	NA	frameshift	c.818delA	p.Asn273fs
2:25248075:TC:T	NA	frameshift	c.816delG	p.Asn273fs
2:25248076:CT:C	NA	frameshift	c.815delA	p.Lys272fs
2:25248078:T:A	NA	stop_gained	c.814A>T	p.Lys272*
2:25248079:GT:G	NA	frameshift	c.812delA	p.Asp271fs
2:25248080:T:TC	NA	frameshift	c.811dupG	p.Asp271fs
2:25248080:TC:T	NA	frameshift	c.811delG	p.Asp271fs
2:25248080:TCCC CAGCATCGGA:T	NA	frameshift	c.799_811delTCCG ATGCTGGGG	p.Ser267fs
2:25248083:CCAG CATCGGACCCAC GGGCTCAGG:C	NA	frameshift	c.784_808delCCTG AGCCCGTGGGTCC GATGCTG	p.Pro262fs
2:25248085:AG:A	NA	frameshift	c.806delC	p.Ala269fs
2:25248085:AGC ATCGGACCCACG GGCTCAG:A	NA	frameshift	c.785_806delCTGA GCCCCGTGGGTCC GATGC	p.Pro262fs
2:25248086:G:GC	NA	frameshift	c.805dupG	p.Ala269fs
2:25248087:C:CA	NA	frameshift	c.804dupT	p.Ala269fs
2:25248087:C:CT	NA	frameshift	c.804_805insA	p.Ala269fs
2:25248088:ATC: A	NA	frameshift	c.802_803delGA	p.Asp268fs
2:25248088:ATCG GACCC:A	NA	frameshift	c.796_803delGGGT CCGA	p.Gly266fs
2:25248089:T:TC	NA	frameshift	c.802dupG	p.Asp268fs

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2:25248090:C:CG	NA	frameshift	c.801dupC	p.Asp268fs
2:25248090:CG:C	NA	frameshift	c.801delC	p.Asp268fs
2:25248092:G:GA	NA	frameshift	c.799dupT	p.Ser267fs
2:25248092:GA:G	NA	frameshift	c.799delT	p.Ser267fs
2:25248092:GAC: G	NA	frameshift	c.798_799delGT	p.Ser267fs
2:25248092:GACC CC:G	NA	frameshift	c.795_799delGGGG T	p.Gly266fs
2:25248093:A:AC	NA	frameshift	c.798dupG	p.Ser267fs
2:25248093:AC:A	NA	frameshift	c.798delG	p.Ser267fs
2:25248096:CCAC GGGCT:C	NA	frameshift	c.788_795delAGCC CGTG	p.Glu263fs
2:25248098:AC:A	NA	frameshift	c.793delG	p.Val265fs
2:25248098:ACG GGC:A	NA	frameshift	c.789_793delGCCC G	p.Glu263fs
2:25248098:ACG GGCTCAGG:A	NA	frameshift	c.784_793delCCTG AGCCCG	p.Pro262fs
2:25248099:CG:C	NA	frameshift	c.792delC	p.Val265fs
2:25248099:CGG GCTCAG:C	NA	frameshift	c.785_792delCTGA GCCC	p.Pro262fs
2:25248100:G:GT	NA	frameshift	c.791_792insA	p.Val265fs
2:25248102:G:GC T	NA	frameshift	c.788_789dupAG	p.Pro264fs
2:25248102:GC:G	NA	frameshift	c.789delG	p.Glu263fs
2:25248103:C:CT CAGGCGTGGTAG	NA	frameshift	c.776_788dupCTAC CACGCCTGA	p.Glu263fs
2:25248103:CT:C	NA	frameshift	c.788delA	p.Glu263fs
2:25248104:TC:T	NA	frameshift	c.787delG	p.Glu263fs
2:25248105:C:A	NA	stop_gained	c.787G>T	p.Glu263*
2:25248105:CAG: C	NA	frameshift	c.785_786delCT	p.Pro262fs
2:25248106:AG:A	NA	frameshift	c.785delC	p.Pro262fs
2:25248110:G:GT	NA	frameshift	c.781dupA	p.Thr261fs
2:25248110:GT:G	NA	frameshift	c.781delA	p.Thr261fs

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2:25248111:T:TG	NA	frameshift	c.780dupC	p.Thr261fs
2:25248111:TG:T	NA	frameshift	c.780delC	p.Thr261fs
2:25248111:TGGT A:T	NA	frameshift	c.777_780delTACC	p.Thr260fs
2:25248113:GT:G	NA	frameshift	c.778delA	p.Thr260fs
2:25248113:GTA: G	NA	frameshift	c.777_778delTA	p.Thr260fs
2:25248116:G:GC	NA	frameshift	c.775dupG	p.Ala259fs
2:25248116:GC:G	NA	frameshift	c.775delG	p.Ala259fs
2:25248117:C:CTT A	NA	stop_gained	c.774_775insTAA	p.Val258_Ala259i nsTer
2:25248117:CCA: C	NA	frameshift	c.773_774delTG	p.Val258fs
2:25248118:CA:C	NA	frameshift	c.773delT	p.Val258fs
2:25248119:AC:A	NA	frameshift	c.772delG	p.Val258fs
2:25248120:CA:C	NA	frameshift	c.771delT	p.Val258fs
2:25248120:CAGT G:C	NA	frameshift	c.768_771delCACT	p.Thr257fs
2:25248120:CAGT GGGGGATG:C	NA	frameshift	c.761_771delCATC CCCCACT	p.Ala254fs
2:25248121:A:AC C	NA	frameshift	c.770_771insGG	p.Ala259fs
2:25248121:A:AG T	NA	frameshift	c.769_770dupAC	p.Val258fs
2:25248121:AG:A	NA	frameshift	c.770delC	p.Thr257fs
2:25248121:AGT: A	NA	frameshift	c.769_770delAC	p.Thr257fs
2:25248121:AGT GG:A	NA	frameshift	c.767_770delCCAC	p.Pro256fs
2:25248121:AGT GGGGG:A	NA	frameshift	c.764_770delCCCC CAC	p.Ser255fs
2:25248122:GT:G	NA	frameshift	c.769delA	p.Thr257fs
2:25248122:GTG GGGGA:G	NA	frameshift	c.763_769delTCCC CCA	p.Ser255fs

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2:25248123:T:TG	NA	frameshift	c.768dupC	p.Thr257fs
2:25248123:TG:T	rs776931901	frameshift	c.768delC	p.Thr257fs
2:25248128:G:GC	NA	frameshift	c.763_764insG	p.Ser255fs
2:25248129:ATGC GGGGTCAG:A	NA	frameshift	c.752_762delCTGA CCCCGCA	p.Thr251fs
2:25248130:TG:T	NA	frameshift	c.761delC	p.Ala254fs
2:25248132:C:CG	NA	frameshift	c.759dupC	p.Ala254fs
2:25248132:C:CG G	NA	frameshift	c.758_759dupCC	p.Ala254fs
2:25248132:CG:C	NA	frameshift	c.759delC	p.Ala254fs
2:25248132:CGG GGTCAGTGGGCT GCTGCACAGCAG GAGGGCTGG:C	NA	frameshift	c.725_759delCCAG CCCTCTGCTGTGC AGCAGCCCACTGAC CCC	p.Ala242fs
2:25248136:GT:G	NA	frameshift	c.755delA	p.Asp252fs
2:25248138:C:CA	NA	frameshift	c.753dupT	p.Asp252fs
2:25248139:AG:A	NA	frameshift	c.752delC	p.Thr251fs
2:25248140:GT:G	NA	frameshift	c.751delA	p.Thr251fs
2:25248141:TG:T	rs749072422	frameshift	c.750delC	p.Thr251fs
2:25248141:TGG GCTGCTGCACA:T	NA	frameshift	c.738_750delTGTG CAGCAGCCC	p.Val247fs
2:25248141:TGG GCTGCTGCACAGC AGGAG:T	NA	frameshift	c.731_750delCTCC TGCTGTGCAGCAGC CC	p.Pro244fs
2:25248145:CT:C	NA	frameshift	c.746delA	p.Gln249fs
2:25248146:TG:T	NA	frameshift	c.745delC	p.Gln249fs
2:25248146:TGCT GCACAGCAGGAG G:T	NA	frameshift	c.730_745delCCTC CTGCTGTGCAGC	p.Pro244fs
2:25248147:G:A	rs759747476	stop_gained	c.745C>T	p.Gln249*
2:25248147:G:T	NA	missense	c.745C>A	p.Gln249Lys
2:25248149:TGCA CA:T	NA	frameshift	c.738_742delTGTG C	p.Val247fs

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2:25248149:TGCA CAGCAGGAGGG:T	NA	frameshift	c.729_742delCCCT CCTGCTGTGC	p.Ser243fs
2:25248149:TGCA CAGCAGGAGGGC TG:T	NA	frameshift	c.726_742delCAGC CCTCCTGCTGTGC	p.Ser243fs
2:25248150:G:A	rs772648943	stop_gained	c.742C>T	p.Gln248*
2:25248150:G:GC A	NA	frameshift	c.740_741dupTG	p.Gln248fs
2:25248150:GCAC AGCAGGAGGGCT GGCCTCCTCCA:G	NA	frameshift	c.716_741delTGGA GGAGGCCAGCCCTC CTGCTGTG	p.Val239fs
2:25248152:A:AG	NA	frameshift	c.739_740insC	p.Val247fs
2:25248152:AC:A	NA	frameshift	c.739delG	p.Val247fs
2:25248154:A:AG	NA	frameshift	c.737dupC	p.Val247fs
2:25248154:AG:A	NA	frameshift	c.737delC	p.Ala246fs
2:25248154:AGC AGGAG:A	NA	frameshift	c.731_737delCTCC TGC	p.Pro244fs
2:25248155:GC:G	NA	frameshift	c.736delG	p.Ala246fs
2:25248156:C:CA A	NA	frameshift	c.735_736insTT	p.Ala246fs
2:25248156:CAG GAGGGCTGG:C	NA	frameshift	c.725_735delCCAG CCCTCCT	p.Ala242fs
2:25248156:CAG GAGGGCTGGCCT CCT:C	NA	frameshift	c.719_735delAGGA GGCCAGCCCTCCT	p.Glu240fs
2:25248157:AG:A	rs1467732928	frameshift	c.734delC	p.Pro245fs
2:25248157:AGG AGG:A	NA	frameshift	c.730_734delCCTC C	p.Pro244fs
2:25248157:AGG AGGGCTGGCCTCC T:A	NA	frameshift	c.719_734delAGGA GGCCAGCCCTCC	p.Glu240fs
2:25248158:G:GA	NA	frameshift	c.733_734insT	p.Pro245fs
2:25248160:A:AG	NA	frameshift	c.731dupC	p.Pro245fs

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2:25248160:A:AG GGC	NA	frameshift	c.728_731dupGCC C	p.Ala246fs
2:25248160:AG:A	NA	frameshift	c.731delC	p.Pro244fs
2:25248160:AGG GCT:A	NA	frameshift	c.727_731delAGCC C	p.Pro244fs
2:25248161:G:GG GCTGGCCTCCTCC ACCTTCTGAGACT CC	NA	frameshift	c.702_730dupGGA GTCTCAGAAGGTG GAGGAGGCCAGCC	p.Pro244fs
2:25248161:GGG CT:G	rs767439400	frameshift	c.727_730delAGCC	p.Ser243fs
2:25248162:G:GA GGCCAGCCCTCCT	NA	frameshift	c.729_730insAGGA GGGCTGGCCT	p.Pro244fs
2:25248162:GGC: G	NA	frameshift	c.728_729delGC	p.Ser243fs
2:25248163:G:GC TGGCCTCCTCCAC CTTCTGAGACT	NA	frameshift	c.704_728dupAGT CTCAGAAGGTGGA GGAGGCCAG	p.Ser243fs
2:25248163:GC:G	NA	frameshift	c.728delG	p.Ser243fs
2:25248163:GCTG GCCT:G	NA	frameshift	c.722_728delAGGC CAG	p.Glu241fs
2:25248163:GCTG GCCTC:G	NA	frameshift	c.721_728delGAGG CCAG	p.Glu241fs
2:25248164:CT:C	NA	frameshift	c.727delA	p.Ser243fs
2:25248165:T:TG	NA	frameshift	c.726dupC	p.Ser243fs
2:25248165:TG:T	NA	frameshift	c.726delC	p.Ser243fs
2:25248166:GGC CTC:G	NA	frameshift	c.721_725delGAGG C	p.Glu241fs
2:25248166:GGC CTCCTC:G	NA	frameshift	c.718_725delGAGG AGGC	p.Glu240fs
2:25248167:G:GA GGAGGCCAGC	NA	frameshift	c.724_725insGCTG GCCTCCT	p.Ala242fs
2:25248167:G:GA GTAGGCCAGC	NA	frameshift	c.724_725insGCTG GCCTACT	p.Ala242fs

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2:25248167:G:GC CTCCTCCACCTTCT GAGACTCCCCGG GCCCCTGGTTTTTC TTC	NA	frameshift	c.682_724dupGAA GAAAACCAGGGGC CCGGGGAGTCTCA GAAGGTGGAGGAG G	p.Ala242fs
2:25248167:GC:G	NA	frameshift	c.724delG	p.Ala242fs
2:25248167:GCC: G	NA	frameshift	c.723_724delGG	p.Glu241fs
2:25248167:GCCT CCTC:G	NA	frameshift	c.718_724delGAGG AGG	p.Glu240fs
2:25248169:C:CA GGA	NA	frameshift	c.722_723insTCCT	p.Glu241fs
2:25248171:C:A	NA	stop_gained	c.721G>T	p.Glu241*
2:25248171:CCT: C	NA	frameshift	c.719_720delAG	p.Glu240fs
2:25248174:C:A	NA	stop_gained	c.718G>T	p.Glu240*
2:25248175:CACC TTCTGAG:C	NA	frameshift	c.707_716delCTCA GAAGGT	p.Ser236fs
2:25248176:A:AC CTTC	NA	frameshift	c.711_715dupGAA GG	p.Val239fs
2:25248176:AC:A	NA	frameshift	c.715delG	p.Val239fs
2:25248176:ACCT TCTGAGACTCC:A	NA	frameshift	c.702_715delGGAG TCTCAGAAGG	p.Glu235fs
2:25248178:C:CT	NA	frameshift	c.713dupA	p.Val239fs
2:25248178:CT:C	NA	frameshift	c.713delA	p.Lys238fs
2:25248179:TTC:T	NA	frameshift	c.711_712delGA	p.Lys238fs
2:25248179:TTCT GAGAC:T	NA	frameshift	c.705_712delGTCT CAGA	p.Ser236fs
2:25248180:T:A	NA	stop_gained	c.712A>T	p.Lys238*
2:25248180:TC:T	NA	frameshift	c.711delG	p.Lys238fs
2:25248183:G:A	NA	stop_gained	c.709C>T	p.Gln237*
2:25248183:G:GA	NA	frameshift	c.708dupT	p.Gln237fs
2:25248183:GA:G	NA	frameshift	c.708delT	p.Gln237fs

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2:25248183:GAG ACTCCCCGGGCCC CTGGTTTTCTT:G	NA	frameshift	c.683_708delAAGA AAACCAGGGGCCC GGGGAGTCT	p.Glu228fs
2:25248186:AC:A	NA	frameshift	c.705delG	p.Glu235fs
2:25248187:C:CT	NA	frameshift	c.704dupA	p.Ser236fs
2:25248187:CT:C	NA	frameshift	c.704delA	p.Glu235fs
2:25248188:T:TC	rs1301352218	frameshift	c.703dupG	p.Glu235fs
2:25248188:TC:T	NA	frameshift	c.703delG	p.Glu235fs
2:25248189:C:A	NA	stop_gained	c.703G>T	p.Glu235*
2:25248191:C:CT	NA	frameshift	c.700_701insA	p.Gly234fs
2:25248192:C:CG	NA	frameshift	c.699dupC	p.Gly234fs
2:25248192:C:CG G	NA	frameshift	c.698_699dupCC	p.Gly234fs
2:25248192:CG:C		frameshift	c.699delC	p.Glu235fs
2:25248195:G:GC	NA	frameshift	c.696dupG	p.Pro233fs
2:25248195:G:GC CCCTGGT	NA	frameshift	c.689_696dupACC AGGGG	p.Pro233fs
2:25248195:GC:G	rs763740173	frameshift	c.696delG	p.Glu235fs
2:25248195:GCC: G	NA	frameshift	c.695_696delGG	p.Gly232fs
2:25248196:C:CG	NA	frameshift	c.695_696insC	p.Pro233fs
2:25248199:CT:C		frameshift	c.692delA	p.Gln231fs
2:25248200:T:TG	NA	frameshift	c.691dupC	p.Gln231fs
2:25248200:T:TG GTTTTCTTCCCCC	NA	frameshift	c.691_692insGGGG GAAGAAAACC	p.Gln231fs
2:25248200:TG:T	NA	frameshift	c.691delC	p.Gln231fs
2:25248201:G:A	NA	stop_gained	c.691C>T	p.Gln231*
2:25248202:GT:G	NA	frameshift	c.689delA	p.Asn230fs
2:25248203:TTTT C:T	NA	frameshift	c.685_688delGAAA	p.Glu229fs
2:25248206:TC:T	NA	frameshift	c.685delG	p.Glu229fs
2:25248207:C:A	NA	stop_gained	c.685G>T	p.Glu229*
2:25248207:CT:C	NA	frameshift	c.684delA	p.Glu229fs

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2:25248209:TC:T	NA	frameshift	c.682delG	p.Glu228fs
2:25248210:C:A	NA	stop_gained	c.682G>T	p.Glu228*
2:25248211:CA:C	NA	frameshift	c.680delT	p.Val227fs
2:25248213:C:CA GCA	NA	frameshift	c.675_678dupTGCT	p.Val227fs
2:25248213:CA:C	NA	frameshift	c.678delT	p.Val227fs
2:25248214:AG:A	NA	frameshift	c.677delC	p.Ala226fs
2:25248215:G:GC	NA	frameshift	c.676dupG	p.Ala226fs
2:25248215:GC:G	NA	frameshift	c.676delG	p.Ala226fs
2:25248216:C:CA	NA	frameshift	c.675dupT	p.Ala226fs
2:25248216:CA:C	NA	frameshift	c.675delT	p.Asn225fs
2:25248217:AT:A	rs1306866372	frameshift	c.674delA	p.Asn225fs
2:25248219:TC:T	NA	frameshift	c.672delG	p.Met224fs
2:25248220:CA:C	NA	frameshift	c.671delT	p.Met224fs
2:25248221:ATTC C:A	NA	frameshift	c.667_670delGGAA	p.Gly223fs
2:25274943:C:A	NA	stop_gained	c.637G>T	p.Glu213*
2:25274945:CT:C	NA	frameshift	c.634delA	p.Arg212fs
2:25274950:C:T	NA	stop_gained	c.630G>A	p.Trp210*
2:25274951:C:T	NA	stop_gained	c.629G>A	p.Trp210*
2:25274988:T:TG TAGTAGGGGTCCC CCGCCTGGAAGG TGAGCCTCGGCAT GGGCCGCTGAC	NA	frameshift	c.542_591dupGTC AGCGGCCCATGCCG AGGCTCACCTTCCA GGCGGGGGACCCC TACTAC	p.Ile198fs
2:25275009:G:A	NA	stop_gained	c.571C>T	p.Gln191*
2:25275010:GAA GGTGA:G	NA	frameshift	c.563_569delTCAC CTT	p.Leu188fs
2:25275021:T:TCC GTCAG	NA	frameshift	c.558_559insCTGA CGG	p.Arg187fs
2:25275022:C:CG	NA	frameshift	c.557dupC	p.Arg187fs
2:25275036:G:A	NA	stop_gained	c.544C>T	p.Gln182*
2:25275050:TC:T	NA	frameshift	c.529delG	p.Glu177fs

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2:25275056:CCCA AGCCA:C	NA	frameshift	c.516_523delTGGC TTGG	p.Gly173fs
2:25275064:A:AC	NA	frameshift	c.515dupG	p.Gly173fs
2:25275069:G:GT	NA	frameshift	c.510_511insA	p.Arg171fs
2:25275074:CG:C	rs1458590320	frameshift	c.505delC	p.Arg169fs
2:25275076:GC:G	NA	frameshift	c.503delG	p.Gly168fs
2:25275498:G:C	NA	splice_donor	c.492+2C>G	
2:25275512:G:GG ATTCGAT	NA	frameshift	c.472_479dupATC GAATC	p.Met161fs
2:25275541:T:A	NA	stop_gained	c.451A>T	p.Lys151*
2:25275545:T:C	NA	splice_acceptor	c.449-2A>G	
2:25282440:C:T	NA	splice_donor	c.448+1G>A	
2:25282461:CG:C	NA	frameshift	c.427delC	p.Arg143fs
2:25282462:G:A	rs1352259738	stop_gained	c.427C>T	p.Arg143*
2:25282463:GC:G	NA	frameshift	c.425delG	p.Gly142fs
2:25282486:CA:C	NA	frameshift	c.402delT	p.Asn134fs
2:25282487:AT:A	NA	frameshift	c.401delA	p.Asn134fs
2:25282503:G:T	NA	stop_gained	c.386C>A	p.Ser129*
2:25282538:CT:C	NA	frameshift	c.350delA	p.Glu117fs
2:25282544:T:TG GGGGGCAGAAGG GCGGGGCCCCAG CAGA	NA	frameshift	c.344_345insTCTG CTGGGGCCCCGCCC TTCTGCCCCC	p.Ala116fs
2:25282544:TG:T	NA	frameshift	c.344delC	p.Pro115fs
2:25282548:GCCC C:G	NA	frameshift	c.337_340delGGGG	p.Gly113fs
2:25282551:C:CA GAAGGG	NA	frameshift	c.337_338insCCCTT CT	p.Gly113fs
2:25282561:GC:G	NA	frameshift	c.327delG	p.Gln110fs
2:25282576:TC:T	NA	frameshift	c.312delG	p.Ser105fs
2:25282584:T:TG	NA	frameshift	c.304_305insC	p.Glu102fs
2:25282590:TG:T	NA	frameshift	c.298delC	p.Gln100fs
2:25282591:G:A	NA	stop_gained	c.298C>T	p.Gln100*

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2:25282597:C:A	NA	stop_gained	c.292G>T	p.Glu98*
2:25282610:CA:C	NA	frameshift	c.278delT	p.Leu93fs
2:25282634:TG:T	NA	frameshift	c.254delC	p.Ser85fs
2:25282635:G:C	NA	stop_gained	c.254C>G	p.Ser85*
2:25282642:CTGA GTCCTGGGCCATG G:C	NA	frameshift	c.230_246delCCAT GGCCCAGGACTCA	p.Ser77fs
2:25282644:G:C	NA	stop_gained	c.245C>G	p.Ser82*
2:25282649:C:CC CATCCA	NA	frameshift	c.239_240insTGGA TGG	p.Gln80fs
2:25282651:G:A	NA	stop_gained	c.238C>T	p.Gln80*
2:25282661:TG:T	NA	frameshift	c.227delC	p.Pro76fs
2:25282682:AGG: A	NA	frameshift	c.205_206delCC	p.Pro69fs
2:25282690:T:A	NA	stop_gained	c.199A>T	p.Lys67*
2:25300139:C:CG	rs769876640	frameshift	c.176dupC	p.Val60fs
2:25300231:G:A	NA	stop_gained	c.85C>T	p.Gln29*
2:25313912:C:T	NA	splice_donor	c.72+1G>A	
2:25313936:C:CA GCTCTGCTGCGT	NA	frameshift	c.48_49insACGCAG CAGAGCT	p.Ala17fs
2:25313941:G:GC TC	NA	stop_gained	c.43_44insGAG	p.Ser15delinsTer Ala
2:25313949:G:GC GGAGCGGGAGGA	NA	frameshift	c.35_36insTCCTCCC GCTCCG	p.Ser13fs
2:25313949:G:GC GGAGCGGGC	NA	frameshift	c.35_36insGCCCCG TCCG	p.Ser13fs
2:25313953:TC:T	rs1213222712	frameshift	c.31delG	p.Asp11fs
2:25313957:C:CG	NA	frameshift	c.27dupC	p.Gly10fs
2:25313957:CG:C	NA	frameshift	c.27delC	p.Asp11fs
2:25313968:GA:G	NA	frameshift	c.16delT	p.Ser6fs
2:25313976:G:GC CCTCCA	NA	frameshift	c.8_9insTGGAGGG	p.Met4fs
2:25314163:T:A	NA	splice_acceptor	c.-177-2A>T	

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2:25314163:T:G	NA	splice_acceptor	c.-177-2A>C	

Representative ASXL1 somatic mutations include:

20:32366382:A:G	NA	splice_acceptor	c.58-2A>G	
20:32369013:A:G	NA	splice_acceptor	c.144-2A>G	
20:32429899:A:G	NA	splice_acceptor	c.566-2A>G	
20:32431680:G:A	NA	splice_donor	c.979+1G>A	
20:32432900:C:T	NA	stop_gained	c.1000C>T	p.Gln334*
20:32432930:GA:G	NA	frameshift	c.1034delA	p.Lys345fs
20:32432978:G:T	NA	stop_gained	c.1078G>T	p.Gly360*
20:32432981:C:T	NA	stop_gained	c.1081C>T	p.Gln361*
20:32432986:G:A	NA	splice_donor	c.1085+1G>A	
20:32433283:G:A	NA	splice_acceptor	c.1086-1G>A	
20:32433283:G:C	NA	splice_acceptor	c.1086-1G>C	
20:32433283:G:T	rs777661872	splice_acceptor	c.1086-1G>T	
20:32433297:A:T	NA	stop_gained	c.1099A>T	p.Lys367*
20:32433310:T:A	NA	stop_gained	c.1112T>A	p.Leu371*
20:32433312:C:T	rs199846284	stop_gained	c.1114C>T	p.Gln372*
20:32433315:C:T	rs1427299519	stop_gained	c.1117C>T	p.Gln373*
20:32433322:TG:T	NA	frameshift	c.1127delG	p.Gly376fs
20:32433327:C:T	rs1386196069	stop_gained	c.1129C>T	p.Gln377*
20:32433330:G:T	NA	stop_gained	c.1132G>T	p.Glu378*
20:32433333:G:GA	NA	frameshift	c.1136dupA	p.Ala380fs
20:32433333:G:T	NA	stop_gained	c.1135G>T	p.Glu379*
20:32433339:G:T	NA	stop_gained	c.1141G>T	p.Glu381*
20:32433344:CAAAAGT GGCTTGTGTGT:C	NA	frameshift	c.1147_1163 delAAAAGTG GCTTGTGTG T	p.Lys383fs
20:32433345:A:T	NA	stop_gained	c.1147A>T	p.Lys383*
20:32433354:TTG:T	rs886043994	frameshift	c.1162_1163 delGT	p.Val388fs

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20:32433355:T:A	rs772584710	stop_gained	c.1157T>A	p.Leu386*
20:32433355:TGTGTG:T	NA	frameshift	c.1158_1162 delGTGTG	p.Leu386fs
20:32433356:G:GT	NA	frameshift	c.1159dupT	p.Cys387fs
20:32433359:T:A	NA	stop_gained	c.1161T>A	p.Cys387*
20:32433359:T:GTCC	NA	frameshift	c.1162_1165 dupGTCC	p.Pro389fs
20:32433361:T:TC	NA	frameshift	c.1166dupC	p.Gly390fs
20:32433366:G:T	rs1190212499	stop_gained	c.1168G>T	p.Gly390*
20:32433369:G:T	NA	stop_gained	c.1171G>T	p.Glu391*
20:32433372:TCAGTGC GTATA:T	NA	frameshift	c.1178_1188 delTGCGTAT ACAG	p.Val393fs
20:32433373:C:CA	NA	frameshift	c.1176dupA	p.Val393fs
20:32433373:C:G	NA	stop_gained	c.1175C>G	p.Ser392*
20:32433379:G:GT	NA	frameshift	c.1182dupT	p.Ile395fs
20:32433380:T:TA	NA	frameshift	c.1183dupA	p.Ile395fs
20:32433381:A:AT	NA	frameshift	c.1184dupT	p.Gln396fs
20:32433384:C:T	NA	stop_gained	c.1186C>T	p.Gln396*
20:32433396:G:GC	NA	frameshift	c.1200dupC	p.Thr401fs
20:32433408:C:CGAGAT GG	NA	frameshift	c.1212_1218 dupAGATGG G	p.His407fs
20:32433408:C:T	rs373145711	stop_gained	c.1210C>T	p.Arg404*
20:32433415:GGCATT AAGAAAC:G	NA	frameshift	c.1220_1233 delATTTTAA GAAACGC	p.His407fs
20:32433417:CATT GAAACGCTCT:C	NA	frameshift	c.1220_1236 delATTTTAA GAAACGCTC T	p.His407fs
20:32433418:A:AT	NA	frameshift	c.1224dupT	p.Lys409fs
20:32433418:ATT:A	NA	frameshift	c.1223_1224 delTT	p.Phe408fs

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20:32433422:T:TA	NA	frameshift	c.1226dupA	p.Lys410fs
20:32433423:A:T	NA	stop_gained	c.1225A>T	p.Lys409*
20:32433425:G:GA	NA	frameshift	c.1230dupA	p.Arg411fs
20:32433426:A:T	NA	stop_gained	c.1228A>T	p.Lys410*
20:32433430:GCT:G	NA	frameshift	c.1236_1237 delTC	p.Arg413fs
20:32433433:C:CT	NA	frameshift	c.1236dupT	p.Arg413fs
20:32433434:TCGGCCA GA:T	NA	frameshift	c.1237_1244 delCGGCCAG A	p.Arg413fs
20:32433441:G:GA	NA	frameshift	c.1244dupA	p.Asp415fs
20:32433441:GAT:G	NA	frameshift	c.1244_1245 delAT	p.Asp415fs
20:32433447:C:T	rs375215583	stop_gained	c.1249C>T	p.Arg417*
20:32433451:CCA:C	NA	frameshift	c.1254_1255 delCA	p.Arg419fs
20:32433453:A:T	NA	stop_gained	c.1255A>T	p.Arg419*
20:32433464:G:GA	NA	frameshift	c.1268dupA	p.Asn423fs
20:32433468:CTG:C	rs1269374749	frameshift	c.1272_1273 delGT	p.Tyr425fs
20:32433468:CTGTACA AA:C	NA	frameshift	c.1271_1278 delTGTACAA A	p.Leu424fs
20:32433472:AC:A	rs755250567	frameshift	c.1275delC	p.Tyr425fs
20:32433473:C:CA	rs1187950456, rs886042532	frameshift	c.1281dupA	p.Gln428fs
20:32433473:C:G	NA	stop_gained	c.1275C>G	p.Tyr425*
20:32433474:A:T	NA	stop_gained	c.1276A>T	p.Lys426*
20:32433477:A:T	NA	stop_gained	c.1279A>T	p.Lys427*
20:32433479:A:AG	NA	frameshift	c.1281_1282 insG	p.Gln428fs
20:32433480:C:CA	NA	frameshift	c.1283dupA	p.Glu429fs
20:32433480:C:T	rs886041975	stop_gained	c.1282C>T	p.Gln428*

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20:32433480:CAG:C	NA	frameshift	c.1283_1284 delAG	p.Gln428fs
20:32433482:GGAGTCA GAACAAGCAGGGGTTGC TAAGGATGCAAAATCTG TGGCCTCAGATGTTCCCC TCTACAAGGATGGGGAG GCTAA:G	NA	frameshift	c.1287_1366 delGTCAGAA CAAGCAGGG GTTGCTAAG GATGCAAAA TCTGTGGCC TCAGATGTTC CCCTCTACAA GGATGGGGA GGCTAAGA	p.Glu429fs
20:32433483:G:T	NA	stop_gained	c.1285G>T	p.Glu429*
20:32433486:TC:T	NA	frameshift	c.1289delC	p.Ser430fs
20:32433487:C:A	NA	stop_gained	c.1289C>A	p.Ser430*
20:32433487:C:G	NA	stop_gained	c.1289C>G	p.Ser430*
20:32433489:G:T	NA	stop_gained	c.1291G>T	p.Glu431*
20:32433492:C:T	rs764325672	stop_gained	c.1294C>T	p.Gln432*
20:32433495:G:GC	NA	frameshift	c.1298dupC	p.Gly434fs
20:32433508:AGGATGC AAAATCTGTGGCCTCAG ATGTTCCCCTCTACAAGG ATGGGGAGGCTAAGACT GACCCAGCAG:A	NA	frameshift	c.1313_1380 delATGCAAA ATCTGTGGC CTCAGATGTT CCCCTCTACA AGGATGGGG AGGCTAAGA CTGACCCAG CAGGG	p.Asp438fs
20:32433528:TC:T	NA	frameshift	c.1331delC	p.Ser444fs
20:32433529:C:A	rs373126831	stop_gained	c.1331C>A	p.Ser444*
20:32433529:C:G	rs373126831	stop_gained	c.1331C>G	p.Ser444*
20:32433540:C:CATCCT TGTAGAGGGGAACA	NA	frameshift	c.1342_1343 insATCCTTGT	p.Leu448fs

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			AGAGGGGA ACA	
20:32433542:C:CCCCAT CCTTG	NA	frameshift	c.1344_1345 insCCCATCCT TG	p.Tyr449fs
20:32433544:AC:A		frameshift	c.1347delC	p.Tyr449fs
20:32433544:ACAAGGA TGGGGAGG:A	NA	frameshift	c.1348_1361 delAAGGATG GGGAGGC	p.Lys450fs
20:32433545:C:A	NA	stop_gained	c.1347C>A	p.Tyr449*
20:32433545:C:G	NA	stop_gained	c.1347C>G	p.Tyr449*
20:32433551:TGG:T	NA	frameshift	c.1356_1357 delGG	p.Glu453fs
20:32433552:G:GTCCC	NA	frameshift	c.1354_1355 insTCCC	p.Gly452fs
20:32433555:G:T	NA	stop_gained	c.1357G>T	p.Glu453*
20:32433556:AGGCTAA GACTGACCCAGCAG:A	NA	frameshift	c.1363_1382 delAAGACTG ACCCAGCAG GGCT	p.Lys455fs
20:32433559:C:CT	NA	frameshift	c.1362dupT	p.Lys455fs
20:32433560:T:TA	NA	frameshift	c.1364dupA	p.Thr456fs
20:32433561:A:T	rs1292471399	stop_gained	c.1363A>T	p.Lys455*
20:32433566:TG:T	NA	frameshift	c.1369delG	p.Asp457fs
20:32433567:GACCC:G	NA	frameshift	c.1371_1374 delCCCA	p.Asp457fs
20:32433568:AC:A	NA	frameshift	c.1373delC	p.Pro458fs
20:32433568:ACC:A	NA	frameshift	c.1372_1373 delCC	p.Pro458fs
20:32433572:AGCAGG: A	NA	frameshift	c.1377_1381 delAGGGC	p.Gly460fs
20:32433575:A:AG	NA	frameshift	c.1380dupG	p.Leu461fs
20:32433578:GC:G	NA	frameshift	c.1381delC	p.Leu461fs
20:32433579:C:CT	NA	frameshift	c.1382dupT	p.Ser462fs

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20:32433580:TGA:T	NA	frameshift	c.1384_1385 delAG	p.Ser462fs
20:32433583:G:GC	NA	frameshift	c.1386dupC	p.Ser463fs
20:32433584:CAG:C	NA	frameshift	c.1387_1388 delAG	p.His465fs
20:32433593:T:TA	NA	frameshift	c.1395_1396 insA	p.Leu466fs
20:32433601:GCACATC CT:G	NA	frameshift	c.1405_1412 delACATCCT C	p.Thr469fs
20:32433606:T:TC	NA	frameshift	c.1410dupC	p.Ser471fs
20:32433613:CAGCACC CGACCTGG:C	NA	frameshift	c.1418_1431 delCACCCGA CCTGGAG	p.Ala473fs
20:32433616:C:CA	NA	frameshift	c.1419dupA	p.Pro474fs
20:32433620:CGACCT:C	NA	frameshift	c.1424_1428 delACCTG	p.Asp475fs
20:32433625:T:TG	NA	frameshift	c.1429dupG	p.Glu477fs
20:32433627:G:T	rs141346625	stop_gained	c.1429G>T	p.Glu477*
20:32433636:G:T	rs545224250	stop_gained	c.1438G>T	p.Glu480*
20:32433639:TTC:T	NA	frameshift	c.1442_1443 delTC	p.Phe481fs
20:32433643:CAGTTG:C	NA	frameshift	c.1449_1453 delTGAGT	p.Glu484fs
20:32433646:T:TG	NA	frameshift	c.1448_1449 insG	p.Glu484fs
20:32433648:G:T	NA	stop_gained	c.1450G>T	p.Glu484*
20:32433666:ATCCAGG CTGAGCCAGACAAC:A	NA	frameshift	c.1470_1489 delCCAGGCT GAGCCAGAC AACT	p.Gln491fs
20:32433669:C:T	rs1335820343	stop_gained	c.1471C>T	p.Gln491*
20:32433675:G:T	rs1434942289	stop_gained	c.1477G>T	p.Glu493*
20:32433686:C:CT	NA	frameshift	c.1490dupT	p.Leu497fs

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20:32433688:T:TG	NA	frameshift	c.1492dupG	p.Ala498fs
20:32433693:C:CG	NA	frameshift	c.1496dupG	p.Ala500fs
20:32433694:G:GT	NA	frameshift	c.1497dupT	p.Ala500fs
20:32433696:G:GC	NA	frameshift	c.1500dupC	p.Ser501fs
20:32433699:T:TCTGC	NA	frameshift	c.1502_1505 dupCTGC	p.Ser503fs
20:32433702:G:GC	NA	frameshift	c.1505dupC	p.Ser503fs
20:32433703:C:CT	NA	frameshift	c.1505_1506 insT	p.Ser503fs
20:32433713:C:CA	NA	frameshift	c.1516dupA	p.Arg506fs
20:32433713:CAG:C	rs1377319529	frameshift	c.1517_1518 delGA	p.Arg506fs
20:32433714:A:T	NA	stop_gained	c.1516A>T	p.Arg506*
20:32433732:C:T	rs757832294	stop_gained	c.1534C>T	p.Gln512*
20:32433734:GGAAACT GT:G	NA	frameshift	c.1538_1545 delAAACTGT G	p.Glu513fs
20:32433735:G:GA	NA	frameshift	c.1540dupA	p.Thr514fs
20:32433735:G:T	rs763708711	stop_gained	c.1537G>T	p.Glu513*
20:32433739:CTG:C	rs886041485,r s777537805	frameshift	c.1544_1545 delTG	p.Val515fs
20:32433746:T:TCAGGA AC	NA	frameshift	c.1550_1556 dupAGGAAC C	p.Lys520fs
20:32433747:C:T	rs755464186	stop_gained	c.1549C>T	p.Gln517*
20:32433750:G:T	NA	stop_gained	c.1552G>T	p.Glu518*
20:32433762:C:T	rs772017757	stop_gained	c.1564C>T	p.Gln522*
20:32433764:G:GA	NA	frameshift	c.1568dupA	p.Arg524fs
20:32433765:A:T	NA	stop_gained	c.1567A>T	p.Lys523*
20:32433765:AAG:A	NA	frameshift	c.1570_1571 delAG	p.Arg524fs
20:32433770:G:GA	NA	frameshift	c.1575dupA	p.Ser526fs
20:32433774:T:TC	NA	frameshift	c.1578dupC	p.Phe527fs
20:32433780:G:T	rs746873290	stop_gained	c.1582G>T	p.Glu528*

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20:32433783:C:T	rs770914619	stop_gained	c.1585C>T	p.Gln529*
20:32433786:G:GC	NA	frameshift	c.1589dupC	p.Ala531fs
20:32433786:G:GCGGC CTCTGCATC	NA	frameshift	c.1590_1602 dupGGCCTCT GCATCC	p.Phe535fs
20:32433786:G:GCGGC CTCTGCATCCTTTCC	rs1439862866	frameshift	c.1591_1609 dupGCCTCTG CATCCTTTCC CG	p.Glu537fs
20:32433787:C:CGGCCT CT	NA	frameshift	c.1591_1597 dupGCCTCTG	p.Ala533fs
20:32433789:G:GCCTCT GC	NA	frameshift	c.1592_1598 dupCCTCTGC	p.Ser534fs
20:32433790:C:CCTCTG CATCCTTTTCTTTT	NA	frameshift	c.1605_1606 insTCTTTTCT CTGCATCCTT T	p.Pro536fs
20:32433791:CTCTGCAT CCTT:C	NA	frameshift	c.1596_1606 delTGCATCC TTTC	p.Ala533fs
20:32433792:TCTGCATC CTTTCCGAAAAGAAGC CC:T	NA	frameshift	c.1596_1621 delTGCATCC TTTCCGAAA AGAAGCCCC	p.Ser534fs
20:32433793:C:CT	NA	frameshift	c.1596dupT	p.Ala533fs
20:32433794:TGC:T	NA	frameshift	c.1597_1598 delGC	p.Ala533fs
20:32433800:C:CT	NA	frameshift	c.1605dupT	p.Pro536fs
20:32433800:CTT:C	NA	frameshift	c.1604_1605 delTT	p.Phe535fs
20:32433803:T:TCCCGA AAAGAAGC	NA	frameshift	c.1610_1622 dupAAAAGA AGCCCCG	p.Leu542fs

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20:32433805:C:CCGAAA AGAAGCCCCGGCTTGAA GAT	NA	frameshift	c.1610_1634 dupAAAAGA AGCCCCGGC TTGAAGATC G	p.Gln546fs
20:32433806:C:CGAAAA GA	NA	frameshift	c.1609_1615 dupGAAAAG A	p.Lys539fs
20:32433806:CGAAAA:C	NA	frameshift	c.1612_1616 delAAGAA	p.Lys538fs
20:32433807:G:T	rs369425922	stop_gained	c.1609G>T	p.Glu537*
20:32433809:A:AAAGA AGCCCC	NA	frameshift	c.1612_1621 dupAAGAAG CCCC	p.Arg541fs
20:32433811:A:AGAAG	NA	frameshift	c.1614_1617 dupGAAG	p.Pro540fs
20:32433812:G:GA	NA	frameshift	c.1616dupA	p.Pro540fs
20:32433812:G:GAAGC CCC	NA	frameshift	c.1615_1621 dupAAGCCC C	p.Arg541fs
20:32433812:GAA:G	rs1388881234	frameshift	c.1615_1616 delAA	p.Lys539fs
20:32433813:A:T	NA	stop_gained	c.1615A>T	p.Lys539*
20:32433815:G:GC		frameshift	c.1621dupC	p.Arg541fs
20:32433815:GCC:G	NA	frameshift	c.1620_1621 delCC	p.Arg541fs
20:32433816:C:CCCCG	NA	frameshift	c.1619_1622 dupCCCCG	p.Leu542fs
20:32433816:CCCCGG:C	NA	frameshift	c.1620_1624 delCCGGC	p.Arg541fs
20:32433818:CCG:C	NA	frameshift	c.1621_1622 delCG	p.Arg541fs

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20:32433818:CCGGCTT GA:C	NA	frameshift	c.1621_1628 delCCGGCTTG A	p.Leu542fs
20:32433819:C:CGGCTT GA	NA	frameshift	c.1622_1628 dupGGCTTG A	p.Asp544fs
20:32433819:C:CGGCTT GAAGA	NA	frameshift	c.1622_1631 dupGGCTTG AAGA	p.Asp544fs
20:32433823:T:TTGAAG ATCGTCAG	NA	frameshift	c.1627_1639 dupGAAGAT CGTCAGT	p.Ser547fs
20:32433824:T:TGAAGA TC	rs780939938	frameshift	c.1628_1634 dupAAGATC G	p.Gln546fs
20:32433825:G:T	NA	stop_gained	c.1627G>T	p.Glu543*
20:32433826:AAGATCG TC:A	NA	frameshift	c.1631_1638 delATCGTCA G	p.Asp544fs
20:32433827:A:AGATC	NA	frameshift	c.1631_1634 dupATCG	p.Gln546fs
20:32433829:A:AT	NA	frameshift	c.1632dupT	p.Arg545fs
20:32433829:ATCGTCA GTCCT:A	NA	frameshift	c.1633_1643 delCGTCAGT CCTT	p.Arg545fs
20:32433830:T:TG	NA	frameshift	c.1632_1633 insG	p.Arg545fs
20:32433830:TCGTCA:T	NA	frameshift	c.1633_1637 delCGTCA	p.Arg545fs
20:32433831:C:CT	NA	frameshift	c.1633_1634 insT	p.Arg545fs
20:32433832:G:GT	NA	frameshift	c.1635dupT	p.Gln546fs
20:32433834:C:T	rs1264792645	stop_gained	c.1636C>T	p.Gln546*
20:32433837:T:TC	NA	frameshift	c.1641dupC	p.Phe548fs

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20:32433839:C:CT	rs745845768	frameshift	c.1644dupT	p.Arg549fs
20:32433841:T:TA	rs1211392515	frameshift	c.1643_1644 insA	p.Phe548fs
20:32433844:G:GT	NA	frameshift	c.1647dupT	p.Asn550fs
20:32433846:A:AACACA ATTGAAAGTGTTACAC	NA	frameshift	c.1649_1670 dupACACAA TTGAAAGTG TTCACAC	p.Glu558fs
20:32433846:AAC:A	NA	frameshift	c.1652_1653 delCA	p.Thr551fs
20:32433855:G:T	NA	stop_gained	c.1657G>T	p.Glu553*
20:32433879:C:T	NA	stop_gained	c.1681C>T	p.Gln561*
20:32433881:G:GC	NA	frameshift	c.1686dupC	p.Thr563fs
20:32433887:TAA:T	NA	frameshift	c.1691_1692 delAA	p.Lys564fs
20:32433900:A:T	NA	stop_gained	c.1702A>T	p.Lys568*
20:32433913:TCCGGGT A:T	NA	frameshift	c.1716_1719 +3delCCGGG TA	p.Arg573fs
20:32433918:G:A	NA	splice_donor	c.1719+1G>A	
20:32433918:G:C	NA	splice_donor	c.1719+1G>C	
20:32433919:T:A	NA	splice_donor	c.1719+2T>A	
20:32433919:T:C	NA	splice_donor	c.1719+2T>C	
20:32434429:CAGATTC A:C	NA	splice_acceptor	c.1720- 1_1725delG ATTCAA	p.Ile574_Leu576d el
20:32434430:A:G	rs376029425	splice_acceptor	c.1720-2A>G	
20:32434430:A:T	NA	splice_acceptor	c.1720-2A>T	
20:32434431:G:A	NA	splice_acceptor	c.1720-1G>A	
20:32434431:G:C	NA	splice_acceptor	c.1720-1G>C	
20:32434431:G:T	rs1254271466	splice_acceptor	c.1720-1G>T	
20:32434435:C:T	rs747847938	stop_gained	c.1723C>T	p.Gln575*
20:32434442:C:A	NA	stop_gained	c.1730C>A	p.Ser577*
20:32434442:C:G	NA	stop_gained	c.1730C>G	p.Ser577*

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20:32434448:TCAAACC ACCCTGGG:T	NA	frameshift	c.1737_1750 delCAAACCA CCCTGGG	p.Lys580fs
20:32434449:CAAACCA CCCTGGGT:C	NA	frameshift	c.1738_1751 delAAACCAC CCTGGGT	p.Lys580fs
20:32434450:AAACCAC CCTGGGTGGTT:A	NA	frameshift	c.1741_1757 delCCACCCT GGGTGGTTA A	p.Pro581fs
20:32434454:CACCCT:C	NA	frameshift	c.1743_1747 delACCCT	p.Pro582fs
20:32434454:CACCCTG GGTGG:C	NA	frameshift	c.1743_1753 delACCCTGG GTGG	p.Pro582fs
20:32434455:ACCCTGG GT:A	NA	frameshift	c.1744_1751 delCCCTGGG T	p.Pro582fs
20:32434456:CCCTGGG TG:C	NA	frameshift	c.1745_1752 delCCTGGGT G	p.Pro582fs
20:32434457:C:CCTGGG TGGTTAAAGGT	NA	frameshift	c.1747_1762 dupTGGGTG GTTAAAGGT C	p.Gln588fs
20:32434459:TGGGTGG TTAAA:T	NA	frameshift	c.1750_1760 delGTGGTTA AAGG	p.Val584fs
20:32434460:G:A	rs763361634	stop_gained	c.1748G>A	p.Trp583*
20:32434461:G:A	rs1174760074	stop_gained	c.1749G>A	p.Trp583*
20:32434463:T:TAACC	rs768359987	frameshift	c.1751_1752 insAACC	p.Val585fs
20:32434465:G:GTAAA GGTCAGCCCAC	NA	frameshift	c.1757_1772 dupAAGGTC	p.Tyr591fs

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			AGCCCACTT A	
20:32434466:T:TTAAA	NA	frameshift	c.1755_1758 dupTAAA	p.Gly587fs
20:32434467:T:TA	NA	frameshift	c.1758dupA	p.Gly587fs
20:32434468:A:T	NA	stop_gained	c.1756A>T	p.Lys586*
20:32434472:G:GT	NA	frameshift	c.1761dupT	p.Gln588fs
20:32434473:T:TC	NA	frameshift	c.1762dupC	p.Gln588fs
20:32434474:C:CA	NA	frameshift	c.1763dupA	p.Pro589fs
20:32434474:C:T	rs1486082302	stop_gained	c.1762C>T	p.Gln588*
20:32434478:CCA:C	NA	frameshift	c.1768_1769 delAC	p.Thr590fs
20:32434481:C:CT		frameshift	c.1771dupT	p.Tyr591fs
20:32434481:CTT:C	rs774161869	frameshift	c.1770_1771 delTT	p.Tyr591fs
20:32434483:T:TA	rs1491418513, rs762036456	frameshift	c.1772dupA	p.Tyr591fs
20:32434483:T:TAA	NA	frameshift	c.1772_1773 insAA	p.Tyr591fs
20:32434483:T:TAG	NA	frameshift	c.1772_1773 insGA	p.Tyr591fs
20:32434483:TAC:T	NA	frameshift	c.1772_1773 delAC	p.Tyr591fs
20:32434483:TACC:T	NA	stop_gained	c.1773_1775 delCCA	p.Tyr591_Gln592 delinsTer
20:32434483:TACCAG:T	NA	frameshift	c.1773_1777 delCCAGA	p.Gln592fs
20:32434483:TACCAGA TATGCCCC:T	NA	frameshift	c.1772_1785 delACCAGAT ATGCCCC	p.Tyr591fs
20:32434484:ACCAGAT AT:A	NA	frameshift	c.1773_1780 delCCAGATA T	p.Tyr591fs
20:32434485:C:A	rs371369583	stop_gained	c.1773C>A	p.Tyr591*

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20:32434485:C:G	rs371369583	stop_gained	c.1773C>G	p.Tyr591*
20:32434486:C:T	rs951716574	stop_gained	c.1774C>T	p.Gln592*
20:32434488:GA:G	NA	frameshift	c.1777delA	p.Ile593fs
20:32434488:GAT:G	NA	frameshift	c.1779_1780 delAT	p.Ile593fs
20:32434490:T:TA	rs1314081658	frameshift	c.1779dupA	p.Cys594fs
20:32434493:G:GC	NA	frameshift	c.1786dupC	p.Arg596fs
20:32434494:C:A	rs755974145	stop_gained	c.1782C>A	p.Cys594*
20:32434503:C:CATCCC CACCACGG	NA	frameshift	c.1793_1805 dupTCCCCAC CACGGA	p.Glu602fs
20:32434505:T:TCCCCA	NA	frameshift	c.1796_1800 dupCCACC	p.Thr601fs
20:32434509:C:CA	NA	frameshift	c.1798dupA	p.Thr600fs
20:32434514:C:CGGCA	NA	frameshift	c.1804_1805 insCAGG	p.Glu602fs
20:32434516:G:T	rs907795578	stop_gained	c.1804G>T	p.Glu602*
20:32434520:C:CCAACC CCGGCAGGAGGA	NA	frameshift	c.1809_1810 insAACCCCG GCAGGAGGA C	p.Ser604fs
20:32434524:C:CG	NA	frameshift	c.1812_1813 insG	p.Cys605fs
20:32434530:G:GCAGG AGGACTCCGT	NA	frameshift	c.1818_1819 insCAGGAGG ACTCCGT	p.Gly607fs
20:32434543:G:GC	NA	frameshift	c.1833dupC	p.Arg612fs
20:32434543:GCC:G		frameshift	c.1832_1833 delCC	p.Ala611fs
20:32434549:A:AC	NA	frameshift	c.1840dupC	p.Leu614fs
20:32434550:CCCTCGCA GACATTAAAG:C	NA	frameshift	c.1841_1857 delTCGCAGA CATTAAAGC C	p.Leu614fs

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20:32434553:TCG:T	NA	frameshift	c.1843_1844 delGC	p.Ala615fs
20:32434558:GAC:G	NA	frameshift	c.1848_1849 delCA	p.Ile617fs
20:32434560:C:CT	rs767955952	frameshift	c.1848_1849 insT	p.Ile617fs
20:32434560:C:CTA	NA	frameshift	c.1848_1849 insTA	p.Ile617fs
20:32434560:C:CTAAT	NA	frameshift	c.1848_1849 insTAAT	p.Ile617fs
20:32434560:CAT:C		frameshift	c.1849_1850 delAT	p.Ile617fs
20:32434561:A:AT	NA	frameshift	c.1851dupT	p.Lys618fs
20:32434562:T:TA	rs773562079	frameshift	c.1850_1851 insA	p.Lys618fs
20:32434563:T:TA	NA	frameshift	c.1854dupA	p.Ala619fs
20:32434563:TAA:T	NA	frameshift	c.1853_1854 delAA	p.Lys618fs
20:32434567:G:GC	NA	frameshift	c.1858dupC	p.Arg620fs
20:32434567:GCC:G	NA	frameshift	c.1857_1858 delCC	p.Arg620fs
20:32434567:GCCCCGTG CTCTGCAGGTCCGAGGG GCGAGAGGTCAC:G	NA	frameshift	c.1857_1891 delCCGTGCT CTGCAGGTC CGAGGGGCG AGAGGTCAC C	p.Arg620fs
20:32434571:GTGCTC:G	NA	frameshift	c.1863_1867 delTCTGC	p.Leu622fs
20:32434574:CTCTGCA GG:C	NA	frameshift	c.1865_1872 delTGCAGGT C	p.Leu622fs
20:32434579:C:CA	NA	frameshift	c.1868dupA	p.Val624fs

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20:32434579:C:CAGGTC CG	NA	frameshift	c.1871_1877 dupTCCGAG G	p.Ala627fs
20:32434579:C:T	rs111316898	stop_gained	c.1867C>T	p.Gln623*
20:32434580:AGGTCC:A	NA	frameshift	c.1870_1874 delGTCCG	p.Val624fs
20:32434582:G:GT	NA	frameshift	c.1871dupT	p.Arg625fs
20:32434583:T:TA	NA	frameshift	c.1871_1872 insA	p.Arg625fs
20:32434587:A:AG	NA	frameshift	c.1879dupG	p.Ala627fs
20:32434592:C:CGAGA	NA	frameshift	c.1882_1885 dupAGAG	p.Gly629fs
20:32434592:CGA:C	NA	frameshift	c.1884_1885 delAG	p.Gly629fs
20:32434596:AGGTCAC CACTGCCATAGAGAGGC: A	NA	frameshift	c.1887_1909 delTCACCAC TGCCATAGA GAGGCGG	p.Glu635fs
20:32434599:TCACCACT GCCATAGAGAGGCGGC: T	rs766433101	frameshift	c.1900_1922 delAGAGAG GCGGCCACC ACTGCCAT	p.Glu635fs
20:32434608:C:A	rs1356224550	stop_gained	c.1896C>A	p.Cys632*
20:32434610:A:AT	NA	frameshift	c.1899dupT	p.Arg634fs
20:32434611:T:TA	NA	frameshift	c.1900dupA	p.Arg634fs
20:32434611:TAG:T	rs753983192	frameshift	c.1904_1905 delAG	p.Glu635fs
20:32434612:A:T	NA	stop_gained	c.1900A>T	p.Arg634*
20:32434612:AGAGAGG CGGCCACCACTGCCATC: A	rs755053984	frameshift	c.1902_1924 delAGAGGC GGCCACCAC TGCCATCG	p.Glu635fs
20:32434615:G:T	NA	stop_gained	c.1903G>T	p.Glu635*
20:32434621:G:GC	NA	frameshift	c.1911dupC	p.Thr638fs

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20:32434622:CCA:C	NA	frameshift	c.1912_1913 delAC	p.Thr638fs
20:32434623:CACCACT GCCATCGGAGG:C	NA	frameshift	c.1912_1928 delACCACTG CCATCGGAG G	p.Thr638fs
20:32434624:ACCACTG CCATCGGAGGGGGG:A	NA	frameshift	c.1913_1932 delCCACTGC CATCGGAGG GGGG	p.Thr638fs
20:32434624:ACCACTG CCATCGGAGGGGGGGG T:A	NA	frameshift	c.1913_1935 delCCACTGC CATCGGAGG GGGGGGT	p.Thr638fs
20:32434626:C:CACTA	NA	frameshift	c.1917_1918 insAACT	p.Ala640fs
20:32434626:CA:C	NA	frameshift	c.1915delA	p.Thr639fs
20:32434626:CACTGCC ATCGGAGG:C	NA	frameshift	c.1915_1928 delACTGCCA TCGGAGG	p.Thr639fs
20:32434626:CACTGCC ATCGGAGGGGGG:C	NA	frameshift	c.1915_1931 delACTGCCA TCGGAGGGG G	p.Thr639fs
20:32434627:ACTGCCAT CGGAGGG:A	NA	frameshift	c.1916_1929 delCTGCCAT CGGAGGG	p.Thr639fs
20:32434627:ACTGCCAT CGGAGGGGGGGGT:A	NA	frameshift	c.1916_1935 delCTGCCAT CGGAGGGG GGGGT	p.Thr639fs
20:32434628:CTGCCA:C	NA	frameshift	c.1918_1922 delGCCAT	p.Ala640fs

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20:32434628:CTGCCATC GGAG:C	NA	frameshift	c.1917_1927 delTGCCATC GGAG	p.Ala640fs
20:32434628:CTGCCATC GGAGGGG:C	NA	frameshift	c.1917_1930 delTGCCATC GGAGGGG	p.Ala640fs
20:32434629:TGCCATC GGAGG:T	rs1262176022	frameshift	c.1919_1929 delCCATCGG AGGG	p.Ala640fs
20:32434629:TGCCATC GGAGGGGGGGGTGG:T	NA	frameshift	c.1918_1937 delGCCATCG GAGGGGGG GGTGG	p.Ala640fs
20:32434632:CAT:C	NA	frameshift	c.1921_1922 delAT	p.Ile641fs
20:32434632:CATCGGA GG:C	NA	frameshift	c.1921_1928 delATCGGAG G	p.Ile641fs
20:32434635:CGGAG:C	NA	frameshift	c.1926_1929 delAGGG	p.Gly644fs
20:32434635:CGGAGG: C	NA	frameshift	c.1926_1930 delAGGGG	p.Gly644fs
20:32434636:G:T	rs892732207	stop_gained	c.1924G>T	p.Gly642*
20:32434637:G:GA	rs752984377	frameshift	c.1926dupA	p.Gly643fs
20:32434637:G:GT	NA	frameshift	c.1925_1926 insT	p.Gly643fs
20:32434638:A:AG	rs1085307856, rs756958159	frameshift	c.1934dupG	p.Gly646fs
20:32434638:A:AGAGG	NA	frameshift	c.1927_1928 insAGGG	p.Gly643fs
20:32434638:AGGGGG: A	NA	frameshift	c.1930_1934 delGGGGG	p.Gly644fs
20:32434639:G:GA	NA	frameshift	c.1927_1928 insA	p.Gly643fs

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20:32434639:G:GC	NA	frameshift	c.1927_1928 insC	p.Gly643fs
20:32434639:GGGGGG GGT:G	NA	frameshift	c.1930_1937 delGGGGGT GG	p.Gly644fs
20:32434646:G:GT	NA	frameshift	c.1935dupT	p.Gly646fs
20:32434647:TGGCCCG GGTGGAGGTGGCGCGG G:T	NA	frameshift	c.1940_1962 delCGGGTGG AGGTGGCGG CGGGGCC	p.Pro647fs
20:32434649:G:GC	NA	frameshift	c.1940dupC	p.Gly649fs
20:32434649:GCC:G	NA	frameshift	c.1939_1940 delCC	p.Pro647fs
20:32434649:GCCCCGG TGGAGGTGGCGGCGGG GCCA:G	NA	frameshift	c.1940_1965 delCGGGTGG AGGTGGCGG CGGGGCCAC C	p.Pro647fs
20:32434652:CGG:C	NA	frameshift	c.1942_1943 delGG	p.Gly648fs
20:32434652:CGGGTG:C	NA	frameshift	c.1942_1946 delGGTGG	p.Gly648fs
20:32434655:G:GT	NA	frameshift	c.1944dupT	p.Gly649fs
20:32434657:G:T	NA	stop_gained	c.1945G>T	p.Gly649*
20:32434657:GGA:G	NA	frameshift	c.1947_1948 delAG	p.Gly650fs
20:32434662:T:TG	NA	frameshift	c.1952dupG	p.Gly652fs
20:32434662:TGGCGGC GGGGCCACCGATGA:T	NA	frameshift	c.1953_1972 delCGGCGG GGCCACCGA TGAGG	p.Gly652fs
20:32434668:CGGGGCC ACCGATGA:C	NA	frameshift	c.1960_1973 delGCCACCG ATGAGGG	p.Ala654fs

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20:32434670:G:GT	NA	frameshift	c.1958_1959 insT	p.Ala654fs
20:32434672:GCCACCG AT:G	NA	frameshift	c.1961_1968 delCCACCGA T	p.Ala654fs
20:32434675:A:AC	NA	frameshift	c.1965dupC	p.Asp656fs
20:32434676:CCGATG:C	NA	frameshift	c.1965_1969 delCGATG	p.Asp656fs
20:32434680:T:TG	NA	frameshift	c.1969dupG	p.Glu657fs
20:32434680:T:TGAGG	NA	frameshift	c.1973_1976 dupGAGG	p.Gly660fs
20:32434681:G:T	NA	stop_gained	c.1969G>T	p.Glu657*
20:32434684:G:T	NA	stop_gained	c.1972G>T	p.Gly658*
20:32434692:C:CA	NA	frameshift	c.1981dupA	p.Arg661fs
20:32434693:A:T	NA	stop_gained	c.1981A>T	p.Arg661*
20:32434702:AGC:A	NA	frameshift	c.1991_1992 delGC	p.Ser664fs
20:32434702:AGCAGTG GTGAT:A	NA	frameshift	c.1992_2002 delCAGTGGT GATG	p.Ser664fs
20:32434712:A:ATGGTG GTGAGGCC	NA	frameshift	c.2003_2015 dupGTGGTG AGGCCTG	p.Cys672fs
20:32434718:G:GTGAG GCC	NA	frameshift	c.2009_2015 dupAGGCCT G	p.Cys672fs
20:32434721:AGG:A	NA	frameshift	c.2010_2011 delGG	p.Glu670fs
20:32434723:G:GC	NA	frameshift	c.2013dupC	p.Cys672fs
20:32434728:T:A	NA	stop_gained	c.2016T>A	p.Cys672*
20:32434728:T:TGGCCT CCCCTGGGCTCAGGGTG GCCACA	NA	frameshift	c.2020_2021 insTCCCCTG GGCTCAGGG	p.His674fs

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			TGCCACAG GCC	
20:32434731:CCACCCTG A:C	NA	frameshift	c.2020_2027 delCACCTG A	p.His674fs
20:32434732:C:CT	NA	frameshift	c.2020_2021 insT	p.His674fs
20:32434734:CCCTGAG CCCAGGGGAGG:C	NA	frameshift	c.2025_2041 delTGAGCCC AGGGGAGG CC	p.Pro677fs
20:32434737:TGAGCCC AG:T	NA	frameshift	c.2027_2034 delAGCCCAG G	p.Glu676fs
20:32434738:G:T	NA	stop_gained	c.2026G>T	p.Glu676*
20:32434744:A:AG	NA	frameshift	c.2036dupG	p.Gly680fs
20:32434744:AG:A	NA	frameshift	c.2036delG	p.Gly679fs
20:32434747:G:T	NA	stop_gained	c.2035G>T	p.Gly679*
20:32434757:G:GT	NA	frameshift	c.2045_2046 insT	p.Thr683fs
20:32434759:A:AT	NA	frameshift	c.2047_2048 insT	p.Thr683fs
20:32434763:C:CT	NA	frameshift	c.2052dupT	p.Gly685fs
20:32434765:G:T	NA	stop_gained	c.2053G>T	p.Gly685*
20:32434768:A:T	NA	stop_gained	c.2056A>T	p.Lys686*
20:32434769:AGT:A	rs780146542	frameshift	c.2060_2061 delGT	p.Cys687fs
20:32434772:GTACGTC AGATC:G	NA	frameshift	c.2064_2074 delGTCAGAT CTAC	p.Ser689fs
20:32434773:T:A	rs1160049111	stop_gained	c.2061T>A	p.Cys687*
20:32434778:C:CA	NA	frameshift	c.2067dupA	p.Asp690fs
20:32434778:C:G	rs1387681483	stop_gained	c.2066C>G	p.Ser689*
20:32434786:C:T	rs1478929932	stop_gained	c.2074C>T	p.Gln692*

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20:32434789:C:T	rs373221034	stop_gained	c.2077C>T	p.Arg693*
20:32434789:CG:C	NA	frameshift	c.2078delG	p.Arg693fs
20:32434790:GAACAC:G	NA	frameshift	c.2082_2086 delACAAC	p.Gln695fs
20:32434792:AC:A	NA	frameshift	c.2081delC	p.Thr694fs
20:32434795:C:CA	NA	frameshift	c.2085dupA	p.Leu696fs
20:32434795:C:CAACT	NA	frameshift	c.2085_2088 dupACTA	p.Leu697fs
20:32434795:C:CT	NA	frameshift	c.2083_2084 insT	p.Gln695fs
20:32434795:C:T	NA	stop_gained	c.2083C>T	p.Gln695*
20:32434795:CA:C	NA	frameshift	c.2085delA	p.Gln695fs
20:32434797:A:AG	NA	frameshift	c.2085_2086 insG	p.Leu696fs
20:32434797:AC:A	NA	frameshift	c.2086delC	p.Leu696fs
20:32434798:C:CA	NA	frameshift	c.2086_2087 insA	p.Leu696fs
20:32434799:T:TA	rs1284591534	frameshift	c.2088dupA	p.Leu697fs
20:32434800:ACTGCCG C:A	NA	frameshift	c.2091_2097 delGCCGCCT	p.Pro698fs
20:32434804:CCGCCTTA TCCT:C	NA	frameshift	c.2094_2104 delGCCTTAT CCTC	p.Pro699fs
20:32434809:TTATCCTC :T	NA	frameshift	c.2100_2106 delTCCTCTA	p.Tyr700fs
20:32434810:T:TA	NA	frameshift	c.2099dupA	p.Tyr700fs
20:32434810:T:TATCCT CTAAATGGGGAGCATAC CCAGGCC	NA	frameshift	c.2099_2127 dupATCCTCT AAATGGGGA GCATACCCA GGCC	p.Gly710fs
20:32434813:C:CCT	NA	frameshift	c.2104_2105 dupCT	p.Asn703fs
20:32434821:T:TG	NA	frameshift	c.2113dupG	p.Glu705fs

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20:32434821:TG:T	rs778670589	frameshift	c.2113delG	p.Glu705fs
20:32434825:G:T	NA	stop_gained	c.2113G>T	p.Glu705*
20:32434825:GA:G	NA	frameshift	c.2114delA	p.Glu705fs
20:32434827:GCATA:G	NA	frameshift	c.2117_2120 delATAC	p.His706fs
20:32434830:T:TA	NA	frameshift	c.2119dupA	p.Thr707fs
20:32434831:AC:A	NA	frameshift	c.2122delC	p.Gln708fs
20:32434831:ACC:A	NA	frameshift	c.2121_2122 delCC	p.Gln708fs
20:32434832:CCCAGGC CGGAACTG:C	NA	frameshift	c.2124_2137 delGGCCGGA ACTGCCA	p.Gln708fs
20:32434834:C:T	rs755789372	stop_gained	c.2122C>T	p.Gln708*
20:32434839:CG:C	rs772303842	frameshift	c.2129delG	p.Gly710fs
20:32434840:G:T	NA	stop_gained	c.2128G>T	p.Gly710*
20:32434840:GGAACTG CCAT:G	NA	frameshift	c.2130_2139 delAACTGCC ATG	p.Thr711fs
20:32434841:GA:G	NA	frameshift	c.2131delA	p.Thr711fs
20:32434842:AAC:A	NA	frameshift	c.2131_2132 delAC	p.Thr711fs
20:32434843:AC:A	NA	frameshift	c.2132delC	p.Thr711fs
20:32434843:ACTGCCAT G:A	NA	frameshift	c.2132_2139 delCTGCCAT G	p.Thr711fs
20:32434845:TG:T	NA	frameshift	c.2134delG	p.Ala712fs
20:32434845:TGCCATGT CCA:T	NA	frameshift	c.2135_2144 delCCATGTC CAG	p.Ala712fs
20:32434846:GC:G	NA	frameshift	c.2136delC	p.Met713fs
20:32434847:CCA:C	NA	frameshift	c.2136_2137 delCA	p.Met713fs
20:32434851:G:GTC	NA	frameshift	c.2140_2141 dupTC	p.Arg715fs

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20:32434852:TC:T	NA	frameshift	c.2142delC	p.Arg715fs
20:32434852:TCCA:T	NA	stop_gained	c.2141_2143 delCCA	p.Ser714_Arg715 delinsTer
20:32434856:GA:G	NA	frameshift	c.2145delA	p.Ala716fs
20:32434857:AG:A	NA	frameshift	c.2146delG	p.Ala716fs
20:32434858:GC:G	NA	frameshift	c.2147delC	p.Ala716fs
20:32434859:CT:C	NA	frameshift	c.2148delT	p.Arg717fs
20:32434860:T:TAG	NA	frameshift	c.2149_2150 dupAG	p.Arg718fs
20:32434862:GGA:G	NA	frameshift	c.2156_2157 delAG	p.Glu719fs
20:32434862:GGAGA:G	NA	frameshift	c.2154_2157 delAGAG	p.Glu719fs
20:32434863:GA:G	NA	frameshift	c.2152delA	p.Arg718fs
20:32434864:A:T	NA	stop_gained	c.2152A>T	p.Arg718*
20:32434864:AG:A	NA	frameshift	c.2153delG	p.Arg718fs
20:32434867:G:T	NA	stop_gained	c.2155G>T	p.Glu719*
20:32434867:GA:G	NA	frameshift	c.2156delA	p.Glu719fs
20:32434868:A:AG	NA	frameshift	c.2158dupG	p.Asp720fs
20:32434868:AG:A	NA	frameshift	c.2158delG	p.Asp720fs
20:32434874:TG:T	NA	frameshift	c.2163delG	p.Pro722fs
20:32434875:GC:G	NA	frameshift	c.2165delC	p.Pro722fs
20:32434877:CT:C	NA	frameshift	c.2167delT	p.Ser723fs
20:32434878:T:TTCTCA GAGAAGGCAGGTCC	NA	frameshift	c.2170_2171 insAGAGAAG GCAGGTCCT CTC	p.Leu724fs
20:32434878:T:TTTCTCA GAGAAGGCAGGTCC	NA	frameshift	c.2167_2168 insTCTCAGA GAAGGCAGG TCCT	p.Ser723fs
20:32434885:A:T	rs757533853	stop_gained	c.2173A>T	p.Arg725*
20:32434886:GA:G	NA	frameshift	c.2177delA	p.Lys726fs
20:32434888:A:T	rs1415948169	stop_gained	c.2176A>T	p.Lys726*

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20:32434889:AG:A	NA	frameshift	c.2179delG	p.Glu727fs
20:32434891:G:T	rs1264581343	stop_gained	c.2179G>T	p.Glu727*
20:32434891:GA:G	NA	frameshift	c.2180delA	p.Glu727fs
20:32434894:G:T	NA	stop_gained	c.2182G>T	p.Glu728*
20:32434894:GA:G	NA	frameshift	c.2185delA	p.Ser729fs
20:32434898:GC:G	NA	frameshift	c.2187delC	p.Cys730fs
20:32434901:GCCTACTA :G	NA	frameshift	c.2191_2197 delCTACTAC	p.Leu731fs
20:32434902:C:A	rs1297030768	stop_gained	c.2190C>A	p.Cys730*
20:32434903:C:CTA	NA	frameshift	c.2192_2193 dupTA	p.Leu732fs
20:32434904:TACTACA GAGGGCTACAGTTGGAC TC:T	NA	frameshift	c.2195_2219 delTACAGAG GGCTACAGT TGGACTCAC	p.Leu732fs
20:32434906:C:CTA	NA	frameshift	c.2195_2196 dupTA	p.Gln733fs
20:32434908:AC:A	rs777135381	frameshift	c.2197delC	p.Gln733fs
20:32434909:C:T	rs387907078	stop_gained	c.2197C>T	p.Gln733*
20:32434912:A:AGG	NA	frameshift	c.2202_2203 dupGG	p.Ala735fs
20:32434912:AG:A	rs1476818548	frameshift	c.2203delG	p.Ala735fs
20:32434920:AG:A	NA	frameshift	c.2209delG	p.Val737fs
20:32434923:TG:T	NA	frameshift	c.2213delG	p.Gly738fs
20:32434924:G:T	rs1165142818	stop_gained	c.2212G>T	p.Gly738*
20:32434925:GACTC:G	NA	frameshift	c.2216_2219 delTCAC	p.Leu739fs
20:32434927:CT:C	NA	frameshift	c.2216delT	p.Leu739fs
20:32434928:T:TCA	NA	frameshift	c.2219_2220 dupCA	p.Asp741fs
20:32434934:A:ATG	NA	frameshift	c.2223_2224 dupTG	p.Gly742fs
20:32434938:GC:G	NA	frameshift	c.2227delC	p.Leu743fs
20:32434940:TA:T	NA	frameshift	c.2229delA	p.Gly744fs

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20:32434941:A:AG	NA	frameshift	c.2231dupG	p.Asp745fs
20:32434943:GA:G	NA	frameshift	c.2232delA	p.Asp745fs
20:32434946:AT:A	NA	frameshift	c.2235delT	p.Asp745fs
20:32434948:GC:G	NA	frameshift	c.2238delC	p.Ser747fs
20:32434951:TC:T	NA	frameshift	c.2242delC	p.Gln748fs
20:32434954:C:T	rs1202551247	stop_gained	c.2242C>T	p.Gln748*
20:32434954:CA:C	NA	frameshift	c.2244delA	p.Gln748fs
20:32434957:CT:C	NA	frameshift	c.2246delT	p.Leu749fs
20:32434958:TC:T	rs1481246277	frameshift	c.2250delC	p.Val751fs
20:32434962:C:CA	NA	frameshift	c.2250_2251 insA	p.Val751fs
20:32434963:GT:G	NA	frameshift	c.2253delT	p.Ala752fs
20:32434963:GTTGCTCC CAC:G	NA	frameshift	c.2253_2262 delTGCTCCC ACT	p.Ala752fs
20:32434965:T:TA	NA	frameshift	c.2253_2254 insA	p.Ala752fs
20:32434965:T:TG	rs1214641052	frameshift	c.2254dupG	p.Ala752fs
20:32434965:TG:T	NA	frameshift	c.2254delG	p.Ala752fs
20:32434968:TC:T	NA	frameshift	c.2259delC	p.Thr754fs
20:32434968:TCCCACTG GGGACCAGCCATG:T	NA	frameshift	c.2259_2278 delCACTGGG GACCAGCCA TGCC	p.Thr754fs
20:32434970:C:CCCACT GGGAG	NA	frameshift	c.2259_2260 insCAGTGGG AGC	p.Thr754fs
20:32434970:CCACTG:C	NA	frameshift	c.2259_2263 delCACTG	p.Thr754fs
20:32434970:CCACTGG GGACCAGCCA:C	NA	frameshift	c.2259_2274 delCACTGGG GACCAGCCA	p.Thr754fs

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20:32434971:C:CCAGTG GGAGCA	NA	frameshift	c.2259_2260 insCAGTGGG AGCA	p.Thr754fs
20:32434971:CA:C	NA	frameshift	c.2260delA	p.Thr754fs
20:32434972:AC:A	NA	frameshift	c.2261delC	p.Thr754fs
20:32434973:CT:C	NA	frameshift	c.2262delT	p.Asp756fs
20:32434974:TG:T	NA	frameshift	c.2266delG	p.Asp756fs
20:32434979:AC:A	rs1449831583	frameshift	c.2269delC	p.Gln757fs
20:32434981:C:T	rs779078826	stop_gained	c.2269C>T	p.Gln757*
20:32434985:CA:C	NA	frameshift	c.2274delA	p.Cys759fs
20:32434986:A:AT		frameshift	c.2275dupT	p.Cys759fs
20:32434986:A:ATG	NA	frameshift	c.2275_2276 dupTG	p.Gln760fs
20:32434988:GCCAGGC CTTGCCC:G	NA	frameshift	c.2278_2290 delCAGGCCT TGCCCC	p.Gln760fs
20:32434989:C:A	NA	stop_gained	c.2277C>A	p.Cys759*
20:32434990:C:T	rs1167715259	stop_gained	c.2278C>T	p.Gln760*
20:32434990:CA:C	NA	frameshift	c.2279delA	p.Gln760fs
20:32434993:GC:G	NA	frameshift	c.2283delC	p.Leu762fs
20:32434995:C:CT	NA	frameshift	c.2285dupT	p.Leu762fs
20:32434997:T:A	NA	stop_gained	c.2285T>A	p.Leu762*
20:32434998:GC:G	NA	frameshift	c.2290delC	p.Leu764fs
20:32434998:GCC:G	NA	frameshift	c.2289_2290 delCC	p.Leu764fs
20:32435003:T:TA	NA	frameshift	c.2292dupA	p.Leu765fs
20:32435004:AC:A	NA	frameshift	c.2293delC	p.Leu765fs
20:32435004:ACTGTCCT C:A	NA	frameshift	c.2294_2301 delTGTCTC C	p.Leu765fs
20:32435004:ACTGTCCT CCCAAACCTCAGTAG:A	NA	frameshift	c.2296_2317 delTCCTCCC AAACCTCAG TAGCTG	p.Ser766fs

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20:32435005:C:CT	NA	frameshift	c.2294dupT	p.Ser766fs
20:32435005:CT:C	NA	frameshift	c.2294delT	p.Leu765fs
20:32435008:TC:T	rs1315414366	frameshift	c.2298delC	p.Ser767fs
20:32435011:TC:T	NA	frameshift	c.2302delC	p.Gln768fs
20:32435014:C:T	rs770762273	stop_gained	c.2302C>T	p.Gln768*
20:32435014:CA:C	NA	frameshift	c.2305delA	p.Thr769fs
20:32435021:C:G	NA	stop_gained	c.2309C>G	p.Ser770*
20:32435023:GT:G	NA	frameshift	c.2312delT	p.Val771fs
20:32435025:A:AGCTGA GAGATTAGTGGAGCAGC CTCAGTT	NA	frameshift	c.2316_2344 dupTGAGAG ATTAGTGGA GCAGCCTCA GTTGC	p.His782fs
20:32435028:TGA:T	NA	frameshift	c.2321_2322 delGA	p.Arg774fs
20:32435029:G:GA	NA	frameshift	c.2318dupA	p.Arg774fs
20:32435029:G:T	rs759218892	stop_gained	c.2317G>T	p.Glu773*
20:32435032:A:T	NA	stop_gained	c.2320A>T	p.Arg774*
20:32435034:AT:A	rs765327792	frameshift	c.2324delT	p.Leu775fs
20:32435036:T:A	rs752263134	stop_gained	c.2324T>A	p.Leu775*
20:32435036:T:G	rs752263134	stop_gained	c.2324T>G	p.Leu775*
20:32435041:G:T	NA	stop_gained	c.2329G>T	p.Glu777*
20:32435044:C:T	NA	stop_gained	c.2332C>T	p.Gln778*
20:32435050:C:T	rs751021760	stop_gained	c.2338C>T	p.Gln780*
20:32435054:T:TG	NA	frameshift	c.2343dupG	p.His782fs
20:32435065:GT:G	NA	frameshift	c.2355delT	p.Arg786fs
20:32435066:T:TTA	NA	frameshift	c.2355_2356 dupTA	p.Arg786fs
20:32435074:G:GA	NA	frameshift	c.2364dupA	p.Cys789fs
20:32435074:G:T	rs1402270258	stop_gained	c.2362G>T	p.Glu788*
20:32435074:GA:G	NA	frameshift	c.2364delA	p.Glu788fs
20:32435075:A:AT	NA	frameshift	c.2363_2364 insT	p.Glu788fs

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20:32435076:ATG:A	NA	frameshift	c.2367_2368 delTG	p.Cys789fs
20:32435078:G:GT	NA	frameshift	c.2367dupT	p.Glu790fs
20:32435078:GT:G	NA	frameshift	c.2367delT	p.Cys789fs
20:32435079:T:A	NA	stop_gained	c.2367T>A	p.Cys789*
20:32435080:G:T	NA	stop_gained	c.2368G>T	p.Glu790*
20:32435081:AG:A	NA	frameshift	c.2370delG	p.Glu790fs
20:32435089:AC:A	NA	frameshift	c.2379delC	p.Thr794fs
20:32435093:CT:C	NA	frameshift	c.2383delT	p.Ser795fs
20:32435095:T:TA	NA	frameshift	c.2383_2384 insA	p.Ser795fs
20:32435095:TC:T	rs752828734	frameshift	c.2385delC	p.Trp796fs
20:32435097:CT:C	NA	frameshift	c.2386delT	p.Trp796fs
20:32435099:G:A	rs770674396	stop_gained	c.2387G>A	p.Trp796*
20:32435100:G:A	NA	stop_gained	c.2388G>A	p.Trp796*
20:32435100:GGA:G	NA	frameshift	c.2389_2390 delGA	p.Glu797fs
20:32435101:G:T	NA	stop_gained	c.2389G>T	p.Glu797*
20:32435104:AG:A	NA	frameshift	c.2393delG	p.Ser798fs
20:32435106:T:TGATGA TGAGGAGG	NA	frameshift	c.2406_2407 insGGATGAT GAGGAG	p.Gln803fs
20:32435108:AT:A	NA	frameshift	c.2397delT	p.Asp799fs
20:32435109:TG:T	NA	frameshift	c.2398delG	p.Asp800fs
20:32435113:G:T	NA	stop_gained	c.2401G>T	p.Glu801*
20:32435113:GA:G	NA	frameshift	c.2402delA	p.Glu801fs
20:32435116:G:T	NA	stop_gained	c.2404G>T	p.Glu802*
20:32435118:G:GCC	NA	frameshift	c.2407_2408 insCC	p.Gln803fs
20:32435118:GC:G	NA	frameshift	c.2407delC	p.Gln803fs
20:32435119:C:T	rs775071544	stop_gained	c.2407C>T	p.Gln803*
20:32435121:AG:A	NA	frameshift	c.2411delG	p.Gly804fs
20:32435124:A:AC	NA	frameshift	c.2415dupC	p.Thr806fs

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20:32435131:GT:G		frameshift	c.2421delT	p.Pro808fs
20:32435133:T:TC	NA	frameshift	c.2423dupC	p.Ala809fs
20:32435133:TC:T	rs763280972	frameshift	c.2423delC	p.Pro808fs
20:32435133:TCC:T	NA	frameshift	c.2422_2423 delCC	p.Pro808fs
20:32435133:TCCTGC:T	NA	frameshift	c.2422_2426 delCCTGC	p.Pro808fs
20:32435140:GA:G	NA	frameshift	c.2429delA	p.Asp810fs
20:32435141:ACAATGG T:A	NA	frameshift	c.2431_2437 delAATGGTC	p.Asn811fs
20:32435142:CAA:C	NA	frameshift	c.2431_2432 delAA	p.Asn811fs
20:32435145:T:TG	NA	frameshift	c.2435dupG	p.Pro813fs
20:32435148:TC:T	NA	frameshift	c.2439delC	p.Ile814fs
20:32435154:TC:T	NA	frameshift	c.2444delC	p.Pro815fs
20:32435156:CG:C	NA	frameshift	c.2445delG	p.Ser816fs
20:32435159:CTCTA:C	NA	frameshift	c.2448_2451 delTCTA	p.Leu817fs
20:32435159:CTCTAG:C	NA	frameshift	c.2449_2453 delCTAGT	p.Leu817fs
20:32435161:C:CT	NA	frameshift	c.2450dupT	p.Val818fs
20:32435161:CT:C	NA	frameshift	c.2450delT	p.Leu817fs
20:32435164:GT:G	NA	frameshift	c.2453delT	p.Val818fs
20:32435165:TG:T	NA	frameshift	c.2456delG	p.Gly819fs
20:32435170:G:GA	NA	frameshift	c.2459dupA	p.Asp820fs
20:32435171:AT:A	NA	frameshift	c.2460delT	p.Asp820fs
20:32435174:A:AT	NA	frameshift	c.2463dupT	p.Thr822fs
20:32435174:ATACATTA GAGAAAGGAACTGGC:A	NA	frameshift	c.2463_2484 delTACATTA GAGAAAGGA ACTGGC	p.Thr822fs
20:32435175:T:TA	rs1221361018	frameshift	c.2464dupA	p.Thr822fs
20:32435176:A:AC	NA	frameshift	c.2465dupC	p.Leu823fs
20:32435178:AT:A	rs767215739	frameshift	c.2468delT	p.Leu823fs

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20:32435179:T:TA	NA	frameshift	c.2467_2468 insA	p.Leu823fs
20:32435180:T:A	NA	stop_gained	c.2468T>A	p.Leu823*
20:32435180:T:G	NA	stop_gained	c.2468T>G	p.Leu823*
20:32435180:T:TA	NA	frameshift	c.2469dupA	p.Glu824fs
20:32435184:GA:G	NA	frameshift	c.2475delA	p.Gly826fs
20:32435187:A:AG	NA	frameshift	c.2477dupG	p.Thr827fs
20:32435187:AG:A	rs1064796100	frameshift	c.2477delG	p.Gly826fs
20:32435188:G:T	rs774018728	stop_gained	c.2476G>T	p.Gly826*
20:32435189:G:GAAC	NA	frameshift	c.2478_2481 dupAACT	p.Gly828fs
20:32435192:CT:C	NA	frameshift	c.2481delT	p.Gly828fs
20:32435193:T:TG	NA	frameshift	c.2483dupG	p.Gln829fs
20:32435197:C:T	rs1013267324	stop_gained	c.2485C>T	p.Gln829*
20:32435205:TGACA:T	NA	frameshift	c.2495_2498 delACAG	p.Asp832fs
20:32435208:CAGTCATC CCACTATGAAGGATCCT GT:C	NA	frameshift	c.2498_2523 delGTCATCC CACTATGAA GGATCCTGT A	p.Ser833fs
20:32435210:GT:G	NA	frameshift	c.2499delT	p.His834fs
20:32435212:C:CTG	NA	frameshift	c.2500_2501 insTG	p.His834fs
20:32435219:CT:C	NA	frameshift	c.2508delT	p.Met837fs
20:32435221:AT:A	NA	frameshift	c.2510delT	p.Met837fs
20:32435222:TG:T	NA	frameshift	c.2511delG	p.Met837fs
20:32435225:AG:A	NA	frameshift	c.2515delG	p.Asp839fs
20:32435233:GTA:G	NA	frameshift	c.2522_2523 delTA	p.Val841fs
20:32435234:TA:T	rs773442382	frameshift	c.2525delA	p.Asn842fs
20:32435238:TG:T	NA	frameshift	c.2527delG	p.Val843fs

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20:32435238:TGTGACC CC:T	NA	frameshift	c.2527_2534 delGTGACCC C	p.Val843fs
20:32435242:A:AC	rs750170870	frameshift	c.2535dupC	p.Ser846fs
20:32435242:AC:A	NA	frameshift	c.2535delC	p.Ser846fs
20:32435248:AGTTCCAC ACCTGAATCCTC:A	NA	frameshift	c.2537_2555 delGTTCCAC ACCTGAATC CTC	p.Ser846fs
20:32435251:TC:T	NA	frameshift	c.2541delC	p.Thr848fs
20:32435252:CCA:C	NA	frameshift	c.2544_2545 delAC	p.Pro849fs
20:32435256:AC:A	NA	frameshift	c.2546delC	p.Pro849fs
20:32435260:G:T	rs914002110	stop_gained	c.2548G>T	p.Glu850*
20:32435263:TC:T	NA	frameshift	c.2553delC	p.Ser852fs
20:32435265:C:CT	NA	frameshift	c.2554dupT	p.Ser852fs
20:32435267:C:G	NA	stop_gained	c.2555C>G	p.Ser852*
20:32435268:A:ACCGAC TGATTGCCTGCAGAACA GAGCATTGATGACGAA TTAGGGCTTGGTGGCTC ATG	NA	frameshift	c.2559_2617 dupGACTGA TTGCCTGCA GAACAGAGC ATTTGATGA CGAATTAGG GCTTGGTGG CTCATGCC	p.Pro873fs
20:32435270:C:CTG	NA	frameshift	c.2558_2559 insTG	p.Thr854fs
20:32435273:CTGAT:C	rs755989932	frameshift	c.2564_2567 delATTG	p.Asp855fs
20:32435274:T:TG	NA	frameshift	c.2563dupG	p.Asp855fs
20:32435274:T:TGATTG CCTGCA	NA	frameshift	c.2565_2575 dupTTGCCTG CAGA	p.Asn859fs

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20:32435275:GATTGCCT GCAGAAC:G	NA	frameshift	c.2565_2578 delTTGCCTG CAGAACA	p.Asp855fs
20:32435277:TTGCC:T	NA	frameshift	c.2569_2572 delCTGC	p.Leu857fs
20:32435283:GCAGAA: G	NA	frameshift	c.2576_2580 delACAGA	p.Asn859fs
20:32435284:C:T	NA	stop_gained	c.2572C>T	p.Gln858*
20:32435286:GA:G	NA	frameshift	c.2576delA	p.Asn859fs
20:32435288:AC:A	NA	frameshift	c.2577delC	p.Asn859fs
20:32435289:CA:C	NA	frameshift	c.2578delA	p.Arg860fs
20:32435289:CAG:C	rs1384553029	frameshift	c.2580_2581 delAG	p.Arg860fs
20:32435290:A:T	NA	stop_gained	c.2578A>T	p.Arg860*
20:32435292:AG:A	NA	frameshift	c.2581delG	p.Ala861fs
20:32435293:GC:G	NA	frameshift	c.2582delC	p.Ala861fs
20:32435294:C:CA	NA	frameshift	c.2583dupA	p.Phe862fs
20:32435299:GA:G	NA	frameshift	c.2588delA	p.Asp863fs
20:32435299:GATGACG AA:G	NA	frameshift	c.2588_2595 delATGACGA A	p.Asp863fs
20:32435305:G:T	rs147895689	stop_gained	c.2593G>T	p.Glu865*
20:32435309:T:G	NA	stop_gained	c.2597T>G	p.Leu866*
20:32435310:AG:A	NA	frameshift	c.2601delG	p.Gly869fs
20:32435316:TG:T	NA	frameshift	c.2606delG	p.Gly869fs
20:32435320:G:GGCTCA TGCCCTCCTATGAGGGA AAGTGATACTAGACAAG AAAACCTT	NA	frameshift	c.2610_2655 dupCTCATGC CCTCCTATGA GGGAAAGTG ATACTAGAC AAGAAAACCT TG	p.Lys886fs
20:32435322:C:CCA	NA	frameshift	c.2610_2611 insCA	p.Ser871fs

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20:32435324:CA:C	rs780091312	frameshift	c.2613delA	p.Cys872fs
20:32435327:G:GC	NA	frameshift	c.2618dupC	p.Pro874fs
20:32435328:C:A	NA	stop_gained	c.2616C>A	p.Cys872*
20:32435328:CCCTCCTA TGA:C	NA	frameshift	c.2617_2626 delCCTCCTAT GA	p.Pro873fs
20:32435338:AG:A	NA	frameshift	c.2629delG	p.Glu877fs
20:32435341:G:GA	NA	frameshift	c.2632dupA	p.Ser878fs
20:32435341:GA:G	NA	frameshift	c.2632delA	p.Ser878fs
20:32435344:AGT:A	NA	frameshift	c.2634_2635 delTG	p.Ser878fs
20:32435349:T:TA	NA	frameshift	c.2638dupA	p.Thr880fs
20:32435350:AC:A	NA	frameshift	c.2639delC	p.Thr880fs
20:32435356:C:T	rs770209084	stop_gained	c.2644C>T	p.Gln882*
20:32435363:AC:A	NA	frameshift	c.2652delC	p.Leu885fs
20:32435366:T:A	NA	stop_gained	c.2654T>A	p.Leu885*
20:32435368:A:T	NA	stop_gained	c.2656A>T	p.Lys886*
20:32435374:A:T	NA	stop_gained	c.2662A>T	p.Lys888*
20:32435375:A:AG	NA	frameshift	c.2665dupG	p.Ala889fs
20:32435380:C:CT	rs1432318140	frameshift	c.2669dupT	p.Val891fs
20:32435385:TTC:T	NA	frameshift	c.2675_2676 delCT	p.Ser892fs
20:32435387:C:CT	NA	frameshift	c.2676dupT	p.Asn893fs
20:32435390:AC:A	NA	frameshift	c.2679delC	p.Asn893fs
20:32435393:GT:G	rs778535367	frameshift	c.2683delT	p.Ser895fs
20:32435395:T:TCTTTG	NA	frameshift	c.2685_2689 dupTTTGC	p.His897fs
20:32435395:TC:T	NA	frameshift	c.2684delC	p.Ser895fs
20:32435396:C:CT	NA	frameshift	c.2687dupT	p.Leu896fs
20:32435405:G:A	NA	stop_gained	c.2693G>A	p.Trp898*
20:32435406:G:A	rs760592730	stop_gained	c.2694G>A	p.Trp898*
20:32435407:A:ATACCC	NA	frameshift	c.2696_2700 dupTACCC	p.Ile901fs

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20:32435407:AT:A	NA	frameshift	c.2696delT	p.Ile899fs
20:32435408:T:TA	NA	frameshift	c.2697dupA	p.Pro900fs
20:32435409:A:AC	NA	frameshift	c.2700dupC	p.Ile901fs
20:32435409:AC:A	NA	frameshift	c.2700delC	p.Ile901fs
20:32435414:TC:T	NA	frameshift	c.2705delC	p.Pro902fs
20:32435418:A:ATCAT	NA	frameshift	c.2708_2709 insATTC	p.Asn904fs
20:32435419:TCGAATG A:T	NA	frameshift	c.2708_2714 delCGAATGA	p.Ser903fs
20:32435421:GA:G	NA	frameshift	c.2711delA	p.Asn904fs
20:32435428:GA:G	NA	frameshift	c.2717delA	p.Glu906fs
20:32435436:G:GA	NA	frameshift	c.2727dupA	p.Gln910fs
20:32435436:GAA:G	NA	frameshift	c.2726_2727 delAA	p.Lys909fs
20:32435440:C:T	NA	stop_gained	c.2728C>T	p.Gln910*
20:32435452:G:T	NA	stop_gained	c.2740G>T	p.Glu914*
20:32435458:A:AT	NA	frameshift	c.2746_2747 insT	p.Arg916fs
20:32435460:AG:A	NA	frameshift	c.2749delG	p.Glu917fs
20:32435462:A:AAC	NA	frameshift	c.2754_2755 dupCA	p.Ile919fs
20:32435462:AAC:A	NA	frameshift	c.2754_2755 delCA	p.Ile919fs
20:32435464:CACAT:C	NA	frameshift	c.2755_2758 delATAC	p.Ile919fs
20:32435468:T:TA	rs771822198	frameshift	c.2757dupA	p.Pro920fs
20:32435474:C:CT	NA	frameshift	c.2763dupT	p.Val922fs
20:32435478:TGAGCC:T	NA	frameshift	c.2767_2771 delGAGCC	p.Glu923fs
20:32435481:G:GC	NA	frameshift	c.2773dupC	p.Gln925fs
20:32435481:GC:G	NA	frameshift	c.2773delC	p.Gln925fs
20:32435483:C:CCCAG	NA	frameshift	c.2772_2775 dupCCAG	p.Val926fs
20:32435485:C:T	rs387907077	stop_gained	c.2773C>T	p.Gln925*

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20:32435487:GGTTGGA GAGGAGT:G	NA	frameshift	c.2777_2789 delTTGGAGA GGAGTG	p.Val926fs
20:32435490:TG:T	NA	frameshift	c.2780delG	p.Gly927fs
20:32435493:A:AG	NA	frameshift	c.2782dupG	p.Glu928fs
20:32435494:G:T	NA	stop_gained	c.2782G>T	p.Glu928*
20:32435499:GTGGGAG AAAGCTGCTCCCACCCCT CC:G	NA	frameshift	c.2790_2815 delGGAGAA AGCTGCTCC CACCCTCCT G	p.Trp930fs
20:32435501:G:A	NA	stop_gained	c.2789G>A	p.Trp930*
20:32435503:G:T	NA	stop_gained	c.2791G>T	p.Glu931*
20:32435505:GA:G	NA	frameshift	c.2796delA	p.Ala933fs
20:32435505:GAAAGCT GCTC:G	NA	frameshift	c.2794_2803 delAAAGCTG CTC	p.Lys932fs
20:32435510:C:CT	NA	frameshift	c.2799dupT	p.Ala934fs
20:32435514:T:TCCCAC	NA	frameshift	c.2806_2810 dupACCCC	p.Ala939fs
20:32435518:A:AAG	NA	frameshift	c.2806_2807 insAG	p.Thr936fs
20:32435521:C:CT	NA	frameshift	c.2809_2810 insT	p.Pro937fs
20:32435521:CCTCCTGC A:C	NA	frameshift	c.2810_2817 delCTCCTGC A	p.Pro937fs
20:32435522:C:CT	NA	frameshift	c.2811dupT	p.Pro938fs
20:32435523:TC:T	NA	frameshift	c.2813delC	p.Pro938fs
20:32435525:CTGCA:C	NA	frameshift	c.2815_2818 delGCAT	p.Ala939fs
20:32435528:CA:C	NA	frameshift	c.2817delA	p.Leu940fs
20:32435529:AT:A	NA	frameshift	c.2819delT	p.Leu940fs

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20:32435529:ATT:A	NA	frameshift	c.2818_2819 delTT	p.Leu940fs
20:32435540:AT:A	NA	frameshift	c.2831delT	p.Leu944fs
20:32435545:ACAGCTG AG:A	NA	frameshift	c.2834_2841 delCAGCTGA G	p.Thr945fs
20:32435551:G:GAGGA	NA	frameshift	c.2840_2843 dupAGGA	p.Leu950fs
20:32435551:G:T	NA	stop_gained	c.2839G>T	p.Glu947*
20:32435558:G:GTCTA	NA	frameshift	c.2847_2850 dupTCTA	p.Asp951fs
20:32435561:T:TA	NA	frameshift	c.2850dupA	p.Asp951fs
20:32435569:C:CT	NA	frameshift	c.2859dupT	p.Asp954fs
20:32435577:CCTTA:C	rs747315609	frameshift	c.2869_2872 delACTT	p.Thr957fs
20:32435580:T:TA	NA	frameshift	c.2869dupA	p.Thr957fs
20:32435581:AC:A	NA	frameshift	c.2870delC	p.Thr957fs
20:32435582:C:CT	NA	frameshift	c.2872dupT	p.Ser958fs
20:32435585:C:A	NA	stop_gained	c.2873C>A	p.Ser958*
20:32435585:C:G	NA	stop_gained	c.2873C>G	p.Ser958*
20:32435587:CT:C	NA	frameshift	c.2876delT	p.Leu959fs
20:32435591:G:A	rs1438791925	stop_gained	c.2879G>A	p.Trp960*
20:32435592:G:A	rs756688220	stop_gained	c.2880G>A	p.Trp960*
20:32435595:TGTGCCAT CTCGAG:T	NA	frameshift	c.2885_2897 delTGCCATC TCGAGG	p.Val962fs
20:32435596:GT:G	NA	frameshift	c.2885delT	p.Val962fs
20:32435605:C:T	rs397515401	stop_gained	c.2893C>T	p.Arg965*
20:32435608:G:T	NA	stop_gained	c.2896G>T	p.Gly966*
20:32435615:GT:G	NA	frameshift	c.2904delT	p.Ser968fs
20:32435617:GA:G	NA	frameshift	c.2906delA	p.Asp969fs
20:32435630:G:GT	NA	frameshift	c.2920dupT	p.Tyr974fs
20:32435630:G:GTT	NA	frameshift	c.2919_2920 dupTT	p.Tyr974fs

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20:32435632:T:TA	NA	frameshift	c.2921dupA	p.Tyr974fs
20:32435633:AC:A	NA	frameshift	c.2922delC	p.Cys975fs
20:32435634:C:A	rs886039722	stop_gained	c.2922C>A	p.Tyr974*
20:32435634:CT:C	NA	frameshift	c.2923delT	p.Cys975fs
20:32435638:C:T	rs776868653	stop_gained	c.2926C>T	p.Gln976*
20:32435641:C:T	rs1366953593	stop_gained	c.2929C>T	p.Gln977*
20:32435649:C:CA	NA	frameshift	c.2938dupA	p.Ile980fs
20:32435653:GA:G	NA	frameshift	c.2945delA	p.Lys982fs
20:32435658:GCTGAAA ATCAACGGAGACTCTGA AGCACTGAGTC:G	NA	frameshift	c.2949_2982 delGAAAATC AACGGAGAC TCTGAAGCA CTGAGTCCT	p.Lys984fs
20:32435666:TC:T	NA	frameshift	c.2955delC	p.Asn986fs
20:32435667:CA:C	NA	frameshift	c.2957delA	p.Asn986fs
20:32435669:AC:A	NA	frameshift	c.2958delC	p.Asn986fs
20:32435670:C:CG	NA	frameshift	c.2960dupG	p.Asp988fs
20:32435670:CG:C	NA	frameshift	c.2960delG	p.Gly987fs
20:32435673:AGACTCT GAAGCACTGAGTCCTCA CG:A	NA	frameshift	c.2963_2987 delACTCTGA AGCACTGAG TCCTCACGG	p.Asp988fs
20:32435675:ACT:A	rs1064796772	frameshift	c.2966_2967 delCT	p.Ser989fs
20:32435680:G:T	NA	stop_gained	c.2968G>T	p.Glu990*
20:32435682:AGCACT:A	NA	frameshift	c.2972_2976 delCACTG	p.Ala991fs
20:32435683:GC:G	NA	frameshift	c.2972delC	p.Ala991fs
20:32435684:CA:C	NA	frameshift	c.2973delA	p.Leu992fs
20:32435689:A:AG	NA	frameshift	c.2978dupG	p.Ser993fs
20:32435695:C:CA	NA	frameshift	c.2984dupA	p.His995fs
20:32435696:ACGGT:A	NA	frameshift	c.2985_2988 delCGGT	p.His995fs
20:32435699:G:GT	NA	frameshift	c.2988dupT	p.Glu997fs

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20:32435701:G:T	NA	stop_gained	c.2989G>T	p.Glu997*
20:32435710:GA:G	NA	frameshift	c.2999delA	p.Asp1000fs
20:32435712:T:TA	NA	frameshift	c.3001dupA	p.Thr1001fs
20:32435716:GC:G	NA	frameshift	c.3006delC	p.Ser1003fs
20:32435717:CCT:C	NA	frameshift	c.3008_3009 delCT	p.Ser1003fs
20:32435724:C:CT	NA	frameshift	c.3015dupT	p.Glu1006fs
20:32435724:CT:C	rs775709283	frameshift	c.3015delT	p.Phe1005fs
20:32435728:GA:G	NA	frameshift	c.3018delA	p.Gly1007fs
20:32435731:G:GCA	NA	frameshift	c.3019_3020 insCA	p.Gly1007fs
20:32435731:GGT:G	NA	frameshift	c.3020_3021 delGT	p.Gly1007fs
20:32435739:C:CA	NA	frameshift	c.3028dupA	p.Thr1010fs
20:32435742:G:GCTGTC CTCCGT	NA	frameshift	c.3030_3031 insCTGTCCTC CGT	p.Glu1011fs
20:32435745:G:GGT	NA	frameshift	c.3034_3035 insTG	p.Asp1012fs
20:32435756:AG:A	NA	frameshift	c.3046delG	p.Ala1016fs
20:32435761:G:GGT	NA	frameshift	c.3049_3050 insGT	p.Asp1017fs
20:32435763:C:CA	NA	frameshift	c.3052dupA	p.Thr1018fs
20:32435764:A:AT	NA	frameshift	c.3052_3053 insT	p.Thr1018fs
20:32435765:CTA:C	NA	frameshift	c.3054_3055 delTA	p.Glu1020fs
20:32435767:A:T	NA	stop_gained	c.3055A>T	p.Arg1019*
20:32435774:CT:C	NA	frameshift	c.3063delT	p.Ala1022fs
20:32435780:TGACAAA GGGATCTTCGGTG:T	NA	frameshift	c.3074_3092 delAGGGATC TTCGGTGGA CAA	p.Lys1025fs
20:32435783:CA:C	NA	frameshift	c.3074delA	p.Lys1025fs

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20:32435790:AT:A	NA	frameshift	c.3079delT	p.Ser1027fs
20:32435793:TTC:T	NA	frameshift	c.3082_3083 delTC	p.Ser1028fs
20:32435795:C:A	rs200702600	stop_gained	c.3083C>A	p.Ser1028*
20:32435796:G:GGTGG ACAAGGATGAGAAACCC AATTGGAACCAATCTGC CCC	NA	frameshift	c.3085_3125 dupGTGGAC AAGGATGAG AAACCCAAT TGGAACCAA TCTGCCCC	p.Leu1043fs
20:32435808:TG:T	NA	frameshift	c.3097delG	p.Glu1033fs
20:32435809:G:T	NA	stop_gained	c.3097G>T	p.Glu1033*
20:32435810:AG:A	NA	frameshift	c.3099delG	p.Lys1034fs
20:32435811:GA:G	NA	frameshift	c.3102delA	p.Lys1034fs
20:32435812:A:AAACCC AATTGGAACCAATCTGC CCCACTGTCCAAGGTG	NA	frameshift	c.3103_3140 dupCCCAATT GGAACCAAT CTGCCCCACT GTCCAAGGT GAA	p.Gly1048fs
20:32435812:A:T	NA	stop_gained	c.3100A>T	p.Lys1034*
20:32435819:AT:A	NA	frameshift	c.3109delT	p.Trp1037fs
20:32435822:G:A	NA	stop_gained	c.3110G>A	p.Trp1037*
20:32435823:G:A	NA	stop_gained	c.3111G>A	p.Trp1037*
20:32435825:ACCAATCT GCC:A	NA	frameshift	c.3117_3126 delATCTGCC CCA	p.Gln1039fs
20:32435827:C:T	rs1221031683	stop_gained	c.3115C>T	p.Gln1039*
20:32435833:GC:G	NA	frameshift	c.3125delC	p.Pro1042fs
20:32435842:T:TC	NA	frameshift	c.3132dupC	p.Lys1045fs
20:32435842:TC:T	NA	frameshift	c.3132delC	p.Lys1045fs
20:32435844:CA:C	NA	frameshift	c.3134delA	p.Lys1045fs
20:32435846:AG:A	NA	frameshift	c.3136delG	p.Val1046fs

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20:32435858:A:ACATG	NA	frameshift	c.3148_3151 dupATGC	p.Arg1051fs
20:32435867:T:TC	NA	frameshift	c.3155_3156 insC	p.Val1053fs
20:32435899:C:CA	NA	frameshift	c.3188dupA	p.Ser1064fs
20:32435899:C:T	rs1311033207	stop_gained	c.3187C>T	p.Gln1063*
20:32435902:AGCTGG:A	NA	frameshift	c.3192_3196 delCTGGG	p.Trp1065fs
20:32435907:G:A	NA	stop_gained	c.3195G>A	p.Trp1065*
20:32435910:GTC:G	NA	frameshift	c.3201_3202 delTC	p.Arg1068fs
20:32435914:C:T	rs764651405	stop_gained	c.3202C>T	p.Arg1068*
20:32436044:T:A	NA	stop_gained	c.3332T>A	p.Leu1111*
20:32436053:GTAGCTT GCCCC:G	NA	frameshift	c.3345_3355 delCTTGCCCC CTAG	p.Ser1115fs
20:32436078:TCC:T	NA	frameshift	c.3367_3368 delCC	p.Pro1123fs
20:32436115:C:T	NA	stop_gained	c.3403C>T	p.Gln1135*
20:32436130:A:T	NA	stop_gained	c.3418A>T	p.Lys1140*
20:32436136:C:T	NA	stop_gained	c.3424C>T	p.Gln1142*
20:32436137:A:AGAGCC ATGCGTAGC	NA	frameshift	c.3433_3434 insCGTAGCG AGCCATG	p.Gly1145fs
20:32436188:GTGGCA: G	NA	frameshift	c.3480_3484 delCATGG	p.Met1161fs
20:32436208:AG:A	NA	frameshift	c.3497delG	p.Ser1166fs
20:32436213:CA:C	NA	frameshift	c.3502delA	p.Ser1168fs
20:32436215:GT:G	NA	frameshift	c.3505delT	p.Ser1169fs
20:32436223:AG:A	NA	frameshift	c.3514delG	p.Ala1172fs
20:32436258:T:A	NA	stop_gained	c.3546T>A	p.Cys1182*
20:32436286:G:T	NA	stop_gained	c.3574G>T	p.Glu1192*
20:32436295:C:T	NA	stop_gained	c.3583C>T	p.Gln1195*
20:32436300:TC:T	NA	frameshift	c.3590delC	p.Pro1197fs

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20:32436303:T:TGGAGC ACCCCA	NA	frameshift	c.3592_3602 dupGGAGCA CCCCA	p.Lys1202fs
20:32436305:GA:G	NA	frameshift	c.3594delA	p.Ala1199fs
20:32436309:AC:A	NA	frameshift	c.3601delC	p.Gln1201fs
20:32436344:A:ACTCC	NA	frameshift	c.3637_3640 dupCTCC	p.His1214fs
20:32436385:A:T	NA	stop_gained	c.3673A>T	p.Lys1225*
20:32436397:ATGGATT CCAAAGAGCAGTTCTCTT CCTTT:A	NA	frameshift	c.3686_3714 delTGGATTC CAAAGAGCA GTTCTCTTCC TTT	p.Met1229fs
20:32436407:AAGAGC: A	NA	frameshift	c.3698_3702 delAGCAG	p.Glu1233fs
20:32436411:GCAGTTCT CTTC:G	NA	frameshift	c.3701_3711 delAGTTCTC TTCC	p.Gln1234fs
20:32436412:C:T	NA	stop_gained	c.3700C>T	p.Gln1234*
20:32436432:T:A	NA	stop_gained	c.3720T>A	p.Cys1240*
20:32436439:C:T	NA	stop_gained	c.3727C>T	p.Gln1243*
20:32436463:C:T	NA	stop_gained	c.3751C>T	p.Gln1251*
20:32436497:GC:G	NA	frameshift	c.3788delC	p.Pro1263fs
20:32436501:AG:A	NA	frameshift	c.3791delG	p.Gly1264fs
20:32436508:CT:C	NA	frameshift	c.3798delT	p.Thr1267fs
20:32436510:T:TA	rs1341638621	frameshift	c.3799dupA	p.Thr1267fs
20:32436523:A:AACGA GGTGTCTCGAGGTAGT AAGAT	NA	frameshift	c.3811_3812 insACGAGGT GTTCTCGAG GTAGTAAGA T	p.Thr1271fs
20:32436523:AC:A	NA	frameshift	c.3812delC	p.Thr1271fs
20:32436527:CT:C	NA	frameshift	c.3816delT	p.Arg1273fs
20:32436530:GT:G	NA	frameshift	c.3821delT	p.Phe1274fs

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20:32436536:C:G	NA	stop_gained	c.3824C>G	p.Ser1275*
20:32436568:C:T	NA	stop_gained	c.3856C>T	p.Gln1286*
20:32436573:AGGTC:A	NA	frameshift	c.3864_3867 delTCGG	p.Arg1289fs
20:32436580:GCC:G	NA	frameshift	c.3870_3871 delCC	p.Leu1291fs
20:32436582:CCT:C	NA	frameshift	c.3871_3872 delCT	p.Leu1291fs
20:32436584:TG:T	NA	frameshift	c.3875delG	p.Gly1292fs
20:32436592:C:T	NA	stop_gained	c.3880C>T	p.Gln1294*
20:32436610:C:T	NA	stop_gained	c.3898C>T	p.Gln1300*
20:32436649:ACC:A	NA	frameshift	c.3939_3940 delCC	p.Leu1314fs
20:32436652:C:CT	NA	frameshift	c.3942dupT	p.Gln1315fs
20:32436655:C:T	NA	stop_gained	c.3943C>T	p.Gln1315*
20:32436656:AGCGCC:A	NA	frameshift	c.3945_3949 delGCGCC	p.Gln1315fs
20:32436674:A:AC	NA	frameshift	c.3965dupC	p.Met1323fs
20:32436680:T:TA	NA	frameshift	c.3968_3969 insA	p.Met1323fs
20:32436681:G:GC	NA	frameshift	c.3971dupC	p.Leu1325fs
20:32436727:G:T	NA	stop_gained	c.4015G>T	p.Gly1339*
20:32436760:C:T	rs763386297	stop_gained	c.4048C>T	p.Gln1350*
20:32436764:C:CATGGA GT	NA	frameshift	c.4052_4053 insATGGAGT	p.Pro1352fs
20:32436772:G:T	NA	stop_gained	c.4060G>T	p.Glu1354*
20:32436777:CTG:C	NA	frameshift	c.4066_4067 delTG	p.Trp1356fs
20:32436780:G:A	NA	stop_gained	c.4068G>A	p.Trp1356*
20:32436795:T:TG	NA	frameshift	c.4084dupG	p.Ala1362fs
20:32436820:G:T	NA	stop_gained	c.4108G>T	p.Glu1370*
20:32436823:A:T	NA	stop_gained	c.4111A>T	p.Lys1371*
20:32436833:T:TG	NA	frameshift	c.4127dupG	p.Pro1377fs
20:32436833:TG:T	NA	frameshift	c.4127delG	p.Gly1376fs

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20:32436848:A:AG	NA	frameshift	c.4138dupG	p.Ala1380fs
20:32436851:CAA:C	NA	frameshift	c.4141_4142 delAA	p.Asn1381fs
20:32436858:CGA:C	NA	frameshift	c.4149_4150 delGA	p.Asn1384fs
20:32436862:AACAGG: A	NA	frameshift	c.4152_4156 delCAGGA	p.Asn1384fs
20:32436884:GTCCCC:G	NA	frameshift	c.4174_4178 delCCCCT	p.Pro1392fs
20:32436885:TC:T	NA	frameshift	c.4177delC	p.Leu1393fs
20:32436944:G:A	NA	stop_gained	c.4232G>A	p.Trp1411*
20:32436944:G:GGA	NA	frameshift	c.4233_4234 dupGA	p.Lys1412fs
20:32436945:G:A	rs1189588116	stop_gained	c.4233G>A	p.Trp1411*
20:32436948:ATT:A	NA	frameshift	c.4237_4238 delTT	p.Leu1413fs
20:32436949:T:TGGGG	NA	frameshift	c.4237_4238 insGGGG	p.Leu1413fs
20:32436950:T:G	NA	stop_gained	c.4238T>G	p.Leu1413*
20:32436950:T:TA	NA	frameshift	c.4239dupA	p.Pro1414fs
20:32436955:C:T	NA	stop_gained	c.4243C>T	p.Arg1415*
20:32436977:G:GAGGC TCAC	NA	frameshift	c.4265_4266 insAGGCTCA C	p.Ser1422fs
20:32436985:CTG:C	rs1463049576	frameshift	c.4274_4275 delTG	p.Leu1425fs
20:32437024:C:T	NA	stop_gained	c.4312C>T	p.Gln1438*
20:32437035:T:G	NA	stop_gained	c.4323T>G	p.Tyr1441*
20:32437101:T:TC	NA	frameshift	c.4392dupC	p.Lys1465fs
20:32437129:C:T	NA	stop_gained	c.4417C>T	p.Gln1473*
20:32437240:C:CT	NA	frameshift	c.4529dupT	p.Lys1511fs
20:32437256:TGTGCCA A:T	NA	frameshift	c.4546_4552 delTGCCAAG	p.Cys1516fs
20:32437257:GT:G	NA	frameshift	c.4546delT	p.Cys1516fs

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20:32437260:C:A	NA	stop_gained	c.4548C>A	p.Cys1516*
20:32437297:G:T	NA	stop_gained	c.4585G>T	p.Gly1529*
20:32437323:C:A	NA	stop_gained	c.4611C>A	p.Cys1537*
20:32437329:GGT:G	NA	frameshift	c.4619_4620 delTG	p.Val1540fs

In some embodiments, the subject's burden of having any one or more somatic mutations in DNMT3A and/or ASXL1 can represent a weighted sum of a plurality of any of the DNMT3A and/or ASXL1 somatic mutations. In some embodiments, the burden is calculated using at least about 2, at least about 3, at least about 4, at least about 5, at least about 10, at least about 20, at least about 30, at least about 40, at least about 50, at least about 60, at least about 70, at least about 80, at least about 100, at least about 120, at least about 150, at least about 200, at least about 250, at least about 300, at least about 400, at least about 500, at least about 1,000, at least about 10,000, at least about 100,000, or at least about or more than 1,000,000 mutations present in or around (up to 10 Mb) the DNMT3A gene and/or the ASXL1 gene where the genetic burden is the number of mutations multiplied by the association estimate with lung cancer for each mutation (e.g., a weighted burden score). This can include any somatic mutations in proximity to the DNMT3A gene and/or the ASXL1 gene (up to 10 Mb around the gene) that show a non-zero association with lung cancer. In some embodiments, when the subject has a burden above a desired threshold score, the subject has an increased risk of developing lung cancer. In some embodiments, when the subject has a burden below a desired threshold score, the subject does not have an increased risk of developing lung cancer.

In some embodiments, the burden may be divided into quintiles, e.g., top quintile, intermediate quintile, and bottom quintile, wherein the bottom quintile of burden corresponds to the lowest risk group and the top quintile of burden corresponds to the highest risk group. In some embodiments, a subject having a greater burden comprises the highest weighted burdens, including, but not limited to the top 10%, top 20%, top 30%, top 40%, or top 50% of burdens from a subject population. In some embodiments, the somatic mutations comprise the somatic mutations having association with lung cancer in the top 10%, top 20%, top 30%, top 40%, or top 50% of p-value range for the association. In some embodiments, each of the identified somatic mutations comprise the somatic mutations having association with lung cancer with p-value of about 10^{-2} , about 10^{-3} , about 10^{-4} , about 10^{-5} , about 10^{-6} , about 10^{-7} ,

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about 10^{-8} , about 10^{-9} , about 10^{-10} , about 10^{-11} , about 10^{-12} , about 10^{-13} , about 10^{-14} , or 10^{-15} . In some embodiments, the identified somatic mutations comprise the somatic mutations having association with lung cancer with p-value of less than 5×10^{-8} . In some embodiments, the identified somatic mutations comprise somatic mutations having association with lung cancer in high-risk subjects as compared to the rest of the reference population with odds ratio (OR) about 1.5 or greater, about 1.75 or greater, about 2.0 or greater, or about 2.25 or greater for the top 20% of the distribution; or about 1.5 or greater, about 1.75 or greater, about 2.0 or greater, about 2.25 or greater, about 2.5 or greater, or about 2.75 or greater. In some embodiments, the OR may range from about 1.0 to about 1.5, from about 1.5 to about 2.0, from about 2.0 to about 2.5, from about 2.5 to about 3.0, from about 3.0 to about 3.5, from about 3.5 to about 4.0, from about 4.0 to about 4.5, from about 4.5 to about 5.0, from about 5.0 to about 5.5, from about 5.5 to about 6.0, from about 6.0 to about 6.5, from about 6.5 to about 7.0, or greater than 7.0. In some embodiments, high-risk subjects comprise subjects having burdens in the top decile, quintile, or tertile in a reference population. The threshold of the burden is determined on the basis of the nature of the intended practical application and the risk difference that would be considered meaningful for that practical application.

In some embodiments, when a subject is identified as having an increased risk of developing lung cancer, the subject can undergo any of the procedures described herein related to lung cancer. In some embodiments, the gene burden can be replaced with a survival analysis whereby carriers of the somatic mutation(s) are examined to determine whether they are more likely or less likely to develop lung cancer over time.

The gene burden analyses described herein can also be used as masks for screening a subject for the risk for developing any of the indications.

The present disclosure also provides methods of detecting the presence or absence of an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and/or a PARP1 variant nucleic acid molecule (i.e., a genomic nucleic acid molecule, an mRNA molecule, or a cDNA molecule produced from an mRNA molecule) in a biological sample from a subject. It is understood that gene sequences within a population and mRNA molecules encoded by such genes can vary due to polymorphisms such as single-nucleotide polymorphisms. The sequences provided herein for the LY75, CD164, and PARP1 variant genomic nucleic acid molecules, LY75, CD164, and PARP1 variant mRNA molecules, and LY75, CD164, and PARP1 variant cDNA molecules are only exemplary sequences. Other sequences for the LY75, CD164, and PARP1

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variant genomic nucleic acid molecules, variant mRNA molecules, and variant cDNA molecules are also possible.

The biological sample can be derived from any cell, tissue, or biological fluid from the subject. The biological sample may comprise any clinically relevant tissue, such as a bone marrow sample, a tumor biopsy, a fine needle aspirate, or a sample of bodily fluid, such as blood, gingival crevicular fluid, plasma, serum, lymph, ascitic fluid, cystic fluid, or urine. In some cases, the sample comprises a buccal swab. The biological sample used in the methods disclosed herein can vary based on the assay format, nature of the detection method, and the tissues, cells, or extracts that are used as the sample. A biological sample can be processed differently depending on the assay being employed. For example, when detecting any an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and/or a PARP1 variant nucleic acid molecule, preliminary processing designed to isolate or enrich the biological sample for the genomic DNA can be employed. A variety of techniques may be used for this purpose. When detecting the level of any LY75, CD164, and/or PARP1 variant mRNA molecule, different techniques can be used enrich the biological sample with mRNA molecules. Various methods to detect the presence or level of an mRNA molecule or the presence of a particular variant genomic DNA locus can be used.

In some embodiments, detecting an LY75 predicted loss-of-function polypeptide in a subject comprises performing a sequence analysis on a biological sample obtained from the subject to determine whether an LY75 genomic nucleic acid molecule in the biological sample, and/or an LY75 mRNA molecule in the biological sample, and/or an LY75 cDNA molecule produced from an mRNA molecule in the biological sample, comprises one or more variations that cause a loss-of-function (partial or complete) or are predicted to cause a loss-of-function (partial or complete).

In some embodiments, detecting a CD164 variant nucleic acid molecule in a subject comprises performing a sequence analysis on a biological sample obtained from the subject to determine whether a CD164 genomic nucleic acid molecule in the biological sample, and/or a CD164 mRNA molecule in the biological sample, and/or a CD164 cDNA molecule produced from an mRNA molecule in the biological sample, comprises one or more variations that cause a loss-of-function (partial or complete) or are predicted to cause a loss-of-function (partial or complete).

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In some embodiments, detecting a PARP1 variant nucleic acid molecule in a subject comprises performing a sequence analysis on a biological sample obtained from the subject to determine whether a PARP1 genomic nucleic acid molecule in the biological sample, and/or a PARP1 mRNA molecule in the biological sample, and/or a PARP1 cDNA molecule produced from an mRNA molecule in the biological sample, comprises one or more variations that cause a loss-of-function (partial or complete) or are predicted to cause a loss-of-function (partial or complete).

In some embodiments, the methods of detecting the presence or absence of an LY75 variant nucleic acid molecule (such as, for example, a genomic nucleic acid molecule, an mRNA molecule, and/or a cDNA molecule produced from an mRNA molecule) in a subject, comprise performing an assay on a biological sample obtained from the subject. The assay determines whether a nucleic acid molecule in the biological sample comprises a particular nucleotide sequence.

In some embodiments, the methods of detecting the presence or absence of a CD164 variant nucleic acid molecules (such as, for example, a genomic nucleic acid molecule, an mRNA molecule, and/or a cDNA molecule produced from an mRNA molecule) in a subject, comprise performing an assay on a biological sample obtained from the subject. The assay determines whether a nucleic acid molecule in the biological sample comprises a particular nucleotide sequence.

In some embodiments, the methods of detecting the presence or absence of a PARP1 variant nucleic acid molecule (such as, for example, a genomic nucleic acid molecule, an mRNA molecule, and/or a cDNA molecule produced from an mRNA molecule) in a subject, comprise performing an assay on a biological sample obtained from the subject. The assay determines whether a nucleic acid molecule in the biological sample comprises a particular nucleotide sequence.

In some embodiments, the biological sample comprises a cell or cell lysate. Such methods can further comprise, for example, obtaining a biological sample from the subject comprising an LY75 genomic nucleic acid molecule or mRNA molecule, a CD164 genomic nucleic acid molecule or mRNA molecule, and/or a PARP1 genomic nucleic acid molecule or mRNA molecule, and if mRNA, optionally reverse transcribing the mRNA into cDNA. Such assays can comprise, for example determining the identity of these positions of the particular LY75 nucleic

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acid molecule, particular CD164 nucleic acid molecule, and/or particular PARP1 nucleic acid molecule. In some embodiments, the method is an *in vitro* method.

In some embodiments, the determining step, detecting step, or sequence analysis comprises sequencing at least a portion of the nucleotide sequence of the LY75 genomic nucleic acid molecule, the LY75 mRNA molecule, or the LY75 cDNA molecule in the biological sample, wherein the sequenced portion comprises one or more variations that cause a loss-of-function (partial or complete) or are predicted to cause a loss-of-function (partial or complete).

In some embodiments, the determining step, detecting step, or sequence analysis comprises sequencing at least a portion of the nucleotide sequence of the CD164 genomic nucleic acid molecule, the CD164 mRNA molecule, or the CD164 cDNA molecule in the biological sample, wherein the sequenced portion comprises one or more variations that cause a loss-of-function (partial or complete) or are predicted to cause a loss-of-function (partial or complete).

In some embodiments, the determining step, detecting step, or sequence analysis comprises sequencing at least a portion of the nucleotide sequence of the PARP1 genomic nucleic acid molecule, the PARP1 mRNA molecule, or the PARP1 cDNA molecule in the biological sample, wherein the sequenced portion comprises one or more variations that cause a loss-of-function (partial or complete) or are predicted to cause a loss-of-function (partial or complete).

In some embodiments, the assay comprises sequencing the entire nucleic acid molecule. In some embodiments, only an LY75 genomic nucleic acid molecule is analyzed. In some embodiments, only an LY75 mRNA is analyzed. In some embodiments, only an LY75 cDNA obtained from LY75 mRNA is analyzed.

In some embodiments, the assay comprises sequencing the entire nucleic acid molecule. In some embodiments, only a CD164 genomic nucleic acid molecule is analyzed. In some embodiments, only a CD164 mRNA is analyzed. In some embodiments, only a CD164 cDNA obtained from CD164 mRNA is analyzed.

In some embodiments, the assay comprises sequencing the entire nucleic acid molecule. In some embodiments, only a PARP1 genomic nucleic acid molecule is analyzed. In some embodiments, only a PARP1 mRNA is analyzed. In some embodiments, only a PARP1 cDNA obtained from PARP1 mRNA is analyzed.

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Alteration-specific polymerase chain reaction techniques can be used to detect mutations such as SNPs in a nucleic acid sequence. Alteration-specific primers can be used because the DNA polymerase will not extend when a mismatch with the template is present.

In some embodiments, the nucleic acid molecule in the sample is mRNA and the mRNA
5 is reverse-transcribed into a cDNA prior to the amplifying step. In some embodiments, the nucleic acid molecule is present within a cell obtained from the subject.

In some embodiments, the assay comprises contacting the biological sample with a primer or probe, such as an alteration-specific primer or alteration-specific probe, that specifically hybridizes to an LY75 variant genomic sequence, variant mRNA sequence, or variant
10 cDNA sequence and not the corresponding LY75 reference sequence under stringent conditions, and determining whether hybridization has occurred.

In some embodiments, the assay comprises contacting the biological sample with a primer or probe, such as an alteration-specific primer or alteration-specific probe, that specifically hybridizes to a CD164 variant genomic sequence, variant mRNA sequence, or
15 variant cDNA sequence and not the corresponding CD164 reference sequence under stringent conditions, and determining whether hybridization has occurred.

In some embodiments, the assay comprises contacting the biological sample with a primer or probe, such as an alteration-specific primer or alteration-specific probe, that specifically hybridizes to a PARP1 variant genomic sequence, variant mRNA sequence, or variant
20 cDNA sequence and not the corresponding PARP1 reference sequence under stringent conditions, and determining whether hybridization has occurred.

In some embodiments, the determining step, detecting step, or sequence analysis comprises: a) amplifying at least a portion of the nucleic acid molecule that encodes the LY75 polypeptide; b) labeling the amplified nucleic acid molecule with a detectable label; c)
25 contacting the labeled nucleic acid molecule with a support comprising an alteration-specific probe; and d) detecting the detectable label.

In some embodiments, the determining step, detecting step, or sequence analysis comprises: a) amplifying at least a portion of the nucleic acid molecule that encodes the CD164 polypeptide; b) labeling the amplified nucleic acid molecule with a detectable label; c)
30 contacting the labeled nucleic acid molecule with a support comprising an alteration-specific probe; and d) detecting the detectable label.

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In some embodiments, the determining step, detecting step, or sequence analysis comprises: a) amplifying at least a portion of the nucleic acid molecule that encodes the PARP1 polypeptide; b) labeling the amplified nucleic acid molecule with a detectable label; c) contacting the labeled nucleic acid molecule with a support comprising an alteration-specific probe; and d) detecting the detectable label.

In some embodiments, the assay comprises RNA sequencing (RNA-Seq). In some embodiments, the assays also comprise reverse transcribing mRNA into cDNA, such as by the reverse transcriptase polymerase chain reaction (RT-PCR).

In some embodiments, the methods utilize probes and primers of sufficient nucleotide length to bind to the target nucleotide sequence and specifically detect and/or identify a polynucleotide comprising an LY75 variant genomic nucleic acid molecule, variant mRNA molecule, or variant cDNA molecule; a CD164 variant genomic nucleic acid molecule, variant mRNA molecule, or variant cDNA molecule; and/or a PARP1 variant genomic nucleic acid molecule, variant mRNA molecule, or variant cDNA molecule. The hybridization conditions or reaction conditions can be determined by the operator to achieve this result. The nucleotide length may be any length that is sufficient for use in a detection method of choice, including any assay described or exemplified herein. Such probes and primers can hybridize specifically to a target nucleotide sequence under high stringency hybridization conditions. Probes and primers may have complete nucleotide sequence identity of contiguous nucleotides within the target nucleotide sequence, although probes differing from the target nucleotide sequence and that retain the ability to specifically detect and/or identify a target nucleotide sequence may be designed by conventional methods. Probes and primers can have about 80%, about 85%, about 90%, about 91%, about 92%, about 93%, about 94%, about 95%, about 96%, about 97%, about 98%, about 99%, or 100% sequence identity or complementarity with the nucleotide sequence of the target nucleic acid molecule.

Illustrative examples of nucleic acid sequencing techniques include, but are not limited to, chain terminator (Sanger) sequencing and dye terminator sequencing. Other methods involve nucleic acid hybridization methods other than sequencing, including using labeled primers or probes directed against purified DNA, amplified DNA, and fixed cell preparations (fluorescence in situ hybridization (FISH)). In some methods, a target nucleic acid molecule may be amplified prior to or simultaneous with detection. Illustrative examples of nucleic acid amplification techniques include, but are not limited to, polymerase chain reaction (PCR), ligase

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chain reaction (LCR), strand displacement amplification (SDA), and nucleic acid sequence based amplification (NASBA). Other methods include, but are not limited to, ligase chain reaction, strand displacement amplification, and thermophilic SDA (tSDA).

In hybridization techniques, stringent conditions can be employed such that a probe or primer will specifically hybridize to its target. In some embodiments, a polynucleotide primer or probe under stringent conditions will hybridize to its target sequence to a detectably greater degree than to other non-target sequences, such as, at least 2-fold, at least 3-fold, at least 4-fold, or more over background, including over 10-fold over background. In some embodiments, a polynucleotide primer or probe under stringent conditions will hybridize to its target nucleotide sequence to a detectably greater degree than to other nucleotide sequences by at least 2-fold. In some embodiments, a polynucleotide primer or probe under stringent conditions will hybridize to its target nucleotide sequence to a detectably greater degree than to other nucleotide sequences by at least 3-fold. In some embodiments, a polynucleotide primer or probe under stringent conditions will hybridize to its target nucleotide sequence to a detectably greater degree than to other nucleotide sequences by at least 4-fold. In some embodiments, a polynucleotide primer or probe under stringent conditions will hybridize to its target nucleotide sequence to a detectably greater degree than to other nucleotide sequences by over 10-fold over background. Stringent conditions are sequence-dependent and will be different in different circumstances.

Appropriate stringency conditions which promote DNA hybridization, for example, 6X sodium chloride/sodium citrate (SSC) at about 45°C., followed by a wash of 2X SSC at 50°C, are known or can be found in *Current Protocols in Molecular Biology*, John Wiley & Sons, N.Y. (1989), 6.3.1-6.3.6. Typically, stringent conditions for hybridization and detection will be those in which the salt concentration is less than about 1.5 M Na⁺ ion, typically about 0.01 to 1.0 M Na⁺ ion concentration (or other salts) at pH 7.0 to 8.3 and the temperature is at least about 30°C for short probes (such as, for example, 10 to 50 nucleotides) and at least about 60°C for longer probes (such as, for example, greater than 50 nucleotides). Stringent conditions may also be achieved with the addition of destabilizing agents such as formamide. Optionally, wash buffers may comprise about 0.1% to about 1% SDS. Duration of hybridization is generally less than about 24 hours, usually about 4 to about 12 hours. The duration of the wash time will be at least a length of time sufficient to reach equilibrium.

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In some embodiments, such isolated nucleic acid molecules comprise or consist of at least about 5, at least about 8, at least about 10, at least about 11, at least about 12, at least about 13, at least about 14, at least about 15, at least about 16, at least about 17, at least about 18, at least about 19, at least about 20, at least about 21, at least about 22, at least about 23, at least about 24, at least about 25, at least about 30, at least about 35, at least about 40, at least about 45, at least about 50, at least about 55, at least about 60, at least about 65, at least about 70, at least about 75, at least about 80, at least about 85, at least about 90, at least about 95, at least about 100, at least about 200, at least about 300, at least about 400, at least about 500, at least about 600, at least about 700, at least about 800, at least about 900, at least about 1000, at least about 2000, at least about 3000, at least about 4000, or at least about 5000

nucleotides. In some embodiments, such isolated nucleic acid molecules comprise or consist of at least about 5, at least about 8, at least about 10, at least about 11, at least about 12, at least about 13, at least about 14, at least about 15, at least about 16, at least about 17, at least about 18, at least about 19, at least about 20, at least about 21, at least about 22, at least about 23, at least about 24, or at least about 25 nucleotides. In some embodiments, the isolated nucleic acid molecules comprise or consist of at least about 18 nucleotides. In some embodiments, the isolated nucleic acid molecules comprise or consists of at least about 15 nucleotides. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 10 to about 35, from about 10 to about 30, from about 10 to about 25, from about 12 to about 30, from about 12 to about 28, from about 12 to about 24, from about 15 to about 30, from about 15 to about 25, from about 18 to about 30, from about 18 to about 25, from about 18 to about 24, or from about 18 to about 22 nucleotides. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 18 to about 30 nucleotides. In some embodiments, the isolated nucleic acid molecules comprise or consist of at least about 15 nucleotides to at least about 35 nucleotides.

In some embodiments, such isolated nucleic acid molecules hybridize to LY75 variant nucleic acid molecules (such as genomic nucleic acid molecules, mRNA molecules, and/or cDNA molecules) under stringent conditions. Such nucleic acid molecules can be used, for example, as probes, primers, alteration-specific probes, or alteration-specific primers as described or exemplified herein, and include, without limitation primers, probes, antisense RNAs, shRNAs, and siRNAs, each of which is described in more detail elsewhere herein, and can be used in any of the methods described herein.

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In some embodiments, such isolated nucleic acid molecules hybridize to CD164 variant nucleic acid molecules (such as genomic nucleic acid molecules, mRNA molecules, and/or cDNA molecules) under stringent conditions. Such nucleic acid molecules can be used, for example, as probes, primers, alteration-specific probes, or alteration-specific primers as described or
5 exemplified herein, and include, without limitation primers, probes, antisense RNAs, shRNAs, and siRNAs, each of which is described in more detail elsewhere herein, and can be used in any of the methods described herein.

In some embodiments, such isolated nucleic acid molecules hybridize to PARP1 variant nucleic acid molecules (such as genomic nucleic acid molecules, mRNA molecules, and/or cDNA
10 molecules) under stringent conditions. Such nucleic acid molecules can be used, for example, as probes, primers, alteration-specific probes, or alteration-specific primers as described or exemplified herein, and include, without limitation primers, probes, antisense RNAs, shRNAs, and siRNAs, each of which is described in more detail elsewhere herein, and can be used in any of the methods described herein.

15 In some embodiments, the isolated nucleic acid molecules hybridize to at least about 15 contiguous nucleotides of a nucleic acid molecule that is at least about 70%, at least about 75%, at least about 80%, at least about 85%, at least about 90%, at least about 95%, at least about 96%, at least about 97%, at least about 98%, at least about 99%, or 100% identical to LY75 variant genomic nucleic acid molecules, LY75 variant mRNA molecules, and/or LY75
20 variant cDNA molecules. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 100 nucleotides, or from about 15 to about 35 nucleotides. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 100 nucleotides. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 35 nucleotides.

25 In some embodiments, the isolated nucleic acid molecules hybridize to at least about 15 contiguous nucleotides of a nucleic acid molecule that is at least about 70%, at least about 75%, at least about 80%, at least about 85%, at least about 90%, at least about 95%, at least about 96%, at least about 97%, at least about 98%, at least about 99%, or 100% identical to CD164 variant genomic nucleic acid molecules, CD164 variant mRNA molecules, and/or CD164
30 variant cDNA molecules. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 100 nucleotides, or from about 15 to about 35 nucleotides. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 100 nucleotides. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 35 nucleotides.

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to about 100 nucleotides. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 35 nucleotides.

In some embodiments, the isolated nucleic acid molecules hybridize to at least about 15 contiguous nucleotides of a nucleic acid molecule that is at least about 70%, at least about 75%, at least about 80%, at least about 85%, at least about 90%, at least about 95%, at least about 96%, at least about 97%, at least about 98%, at least about 99%, or 100% identical to PARP1 variant genomic nucleic acid molecules, PARP1 variant mRNA molecules, and/or PARP1 variant cDNA molecules. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 100 nucleotides, or from about 15 to about 35 nucleotides.

In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 100 nucleotides. In some embodiments, the isolated nucleic acid molecules consist of or comprise from about 15 to about 35 nucleotides.

In some embodiments, the alteration-specific probes and alteration-specific primers comprise DNA. In some embodiments, the alteration-specific probes and alteration-specific primers comprise RNA.

In some embodiments, the probes and primers described herein (including alteration-specific probes and alteration-specific primers) have a nucleotide sequence that specifically hybridizes to any of the nucleic acid molecules disclosed herein, or the complement thereof. In some embodiments, the probes and primers specifically hybridize to any of the nucleic acid molecules disclosed herein under stringent conditions.

In some embodiments, the primers, including alteration-specific primers, can be used in second generation sequencing or high throughput sequencing. In some instances, the primers, including alteration-specific primers, can be modified. In particular, the primers can comprise various modifications that are used at different steps of, for example, Massive Parallel Signature Sequencing (MPSS), Polony sequencing, and 454 Pyrosequencing. Modified primers can be used at several steps of the process, including biotinylated primers in the cloning step and fluorescently labeled primers used at the bead loading step and detection step. Polony sequencing is generally performed using a paired-end tags library wherein each molecule of DNA template is about 135 bp in length. Biotinylated primers are used at the bead loading step and emulsion PCR. Fluorescently labeled degenerate nonamer oligonucleotides are used at the detection step. An adaptor can contain a 5'-biotin tag for immobilization of the DNA library onto streptavidin-coated beads.

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The probes and primers described herein can be used to detect a nucleotide variation within any of the LY75 variant genomic nucleic acid molecules, LY75 variant mRNA molecules, and/or LY75 variant cDNA molecules disclosed herein. The primers described herein can be used to amplify LY75 variant genomic nucleic acid molecules, LY75 variant mRNA molecules, or LY75 variant cDNA molecules, or a fragment thereof.

The probes and primers described herein can also be used to detect a nucleotide variation within any of the CD164 variant genomic nucleic acid molecules, CD164 variant mRNA molecules, and/or CD164 variant cDNA molecules disclosed herein. The primers described herein can be used to amplify CD164 variant genomic nucleic acid molecules, CD164 variant mRNA molecules, or CD164 variant cDNA molecules, or a fragment thereof.

The probes and primers described herein can also be used to detect a nucleotide variation within any of the PARP1 variant genomic nucleic acid molecules, PARP1 variant mRNA molecules, and/or PARP1 variant cDNA molecules disclosed herein. The primers described herein can be used to amplify CD164 variant genomic nucleic acid molecules, PARP1 variant mRNA molecules, or PARP1 variant cDNA molecules, or a fragment thereof.

In the context of the disclosure “specifically hybridizes” means that the probe or primer (such as, for example, the alteration-specific probe or alteration-specific primer) does not hybridize to a nucleic acid sequence encoding an LY75 reference genomic nucleic acid molecule, a CD164 reference genomic nucleic acid molecule, a PARP1 reference genomic nucleic acid molecule, an LY75 reference mRNA molecule, or a CD164 reference mRNA molecule, a PARP1 reference mRNA molecule, an LY75 reference cDNA molecule, a CD164 reference cDNA molecule, and/or PARP reference cDNA molecule.

In some embodiments, the probes (such as, for example, an alteration-specific probe) comprise a label. In some embodiments, the label is a fluorescent label, a radiolabel, or biotin.

The present disclosure also provides supports comprising a substrate to which any one or more of the probes disclosed herein is attached. Solid supports are solid-state substrates or supports with which molecules, such as any of the probes disclosed herein, can be associated. A form of solid support is an array. Another form of solid support is an array detector. An array detector is a solid support to which multiple different probes have been coupled in an array, grid, or other organized pattern. A form for a solid-state substrate is a microtiter dish, such as a standard 96-well type. In some embodiments, a multiwell glass slide can be employed that normally contains one array per well.

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The nucleotide sequence of an LY75 reference genomic nucleic acid molecule is set forth in SEQ ID NO:1 (ENSG00000054219.11 encompassing chr2:159,803,355-159,904,756 in the GRCh38/hg38 human genome assembly). The nucleotide sequence of an LY75 variant genomic nucleic acid molecule is set forth in SEQ ID NO:2 (rs78446341; C70,612T; codon 5 70,611-70,613 CCG to CTG; 101,402 bp). In some embodiments, an LY75 variant genomic nucleic acid molecule is rs147820690 comprising a C>T variation at position chr2:159878663 (GRCh38.p13; NC_000002.12:g.159878663C>T).

The nucleotide sequence of an LY75 reference mRNA molecule is set forth in SEQ ID NO:3 (NM_002349.4; Isoform 1; 6,932 nt; LY75 Segment). The nucleotide sequence of another 10 LY75 reference mRNA molecule is set forth in SEQ ID NO:4 (ENST00000504764.5; Isoform 2; 5,650 nt; LY75-CD302). The nucleotide sequence of another LY75 reference mRNA molecule is set forth in SEQ ID NO:5 (ENST00000505052.1; Isoform 3; 5,482 nt; LY75-CD302). The nucleotide sequence of another LY75 reference mRNA molecule is set forth in SEQ ID NO:6 (NM_001198759.1; Isoform 4; 8,919 nt; LY75-CD302). The nucleotide sequence of another LY75 15 reference mRNA molecule is set forth in SEQ ID NO:7 (NM_001198760.1; Isoform 5; 8,751 nt; LY75-CD302). The nucleotide sequence of another LY75 reference mRNA molecule is set forth in SEQ ID NO:8 (AY184222.1; Isoform 6; 5,622 nt; LY75-CD302). The nucleotide sequence of another LY75 reference mRNA molecule is set forth in SEQ ID NO:9 (AY314006.1; Isoform 7; 5,454 nt; LY75-CD302). The nucleotide sequence of another LY75 reference mRNA molecule is 20 set forth in SEQ ID NO:10 (AB208915.1; Isoform 8; 5,713 nt; LY75-Segment). The nucleotide sequence of another LY75 reference mRNA molecule is set forth in SEQ ID NO:11 (AF011333.1; Isoform 9; 6,928 nt; LY75-Segment). The nucleotide sequence of another LY75 reference mRNA molecule is set forth in SEQ ID NO:12 (AF064827.1; Isoform 10; 5,169 nt; LY75-Segment). The nucleotide sequence of another LY75 reference mRNA molecule is set forth in SEQ ID NO:13 25 (ENST00000263636.4; Isoform 11; 6,886 nt; LY75-Segment).

The nucleotide sequence of an LY75 variant mRNA molecule is set forth in SEQ ID NO:14 (NM_002349.4; Isoform 1; rs78446341; C3,814T; Codon 3,813-3,815 CCG to CUG; 6,932 nt; LY75 Segment). The nucleotide sequence of another LY75 variant mRNA molecule is set forth in SEQ ID NO:15 (ENST00000504764.5; Isoform 2; rs78446341; C3,768T; Codon 3,767- 30 3,769 CCG to CUG; 5,650 nt; LY75-CD302). The nucleotide sequence of another LY75 variant mRNA molecule is set forth in SEQ ID NO:16 (ENST00000505052.1; Isoform 3; rs78446341; C3,768T; Codon 3,767-3,769 CCG to CUG; 5,482 nt; LY75-CD302). The nucleotide sequence of

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another LY75 variant mRNA molecule is set forth in SEQ ID NO:17 (NM_001198759.1; Isoform 4; rs78446341; C3,814T; Codon 3,813-3,815 CCG to CUG; 8,919 nt; LY75-CD302). The nucleotide sequence of another LY75 variant mRNA molecule is set forth in SEQ ID NO:18

(NM_001198760.1; Isoform 5; rs78446341; C3,814T; Codon 3,813-3,815 CCG to CUG; 8,751 nt;

5 LY75-CD302). The nucleotide sequence of another LY75 variant mRNA molecule is set forth in SEQ ID NO:19 (AY184222.1; Isoform 6; rs78446341; C3,740T; Codon 3,739-3,741 CCG to CUG;

5,622 nt; LY75-CD302). The nucleotide sequence of another LY75 variant mRNA molecule is set forth in SEQ ID NO:20 (AY314006.1; Isoform 7; rs78446341; C3,740T; Codon 3,739-3,741 CCG to CUG; 5,454 nt; LY75-CD302). The nucleotide sequence of another LY75 variant mRNA molecule

10 is set forth in SEQ ID NO:21 (AB208915.1; Isoform 8; C2,594T; Codon 2,593-2,595 CCG to CUG; 5,713 nt; LY75-Segment). The nucleotide sequence of another LY75 variant mRNA molecule is

set forth in SEQ ID NO:22 (AF011333.1; Isoform 9; C3,793T; Codon 3,792-3,794 CCG to CUG;

6,928 nt; LY75-Segment). The nucleotide sequence of another LY75 variant mRNA molecule is

set forth in SEQ ID NO:23 (AF064827.1; Isoform 10; C3,740T; Codon 3,739-3,741 CCG to CUG;

15 5,169 nt; LY75-Segment). The nucleotide sequence of another LY75 variant mRNA molecule is set forth in SEQ ID NO:24 (ENST00000263636.4; Isoform 11; C3,768T; Codon 3,767-3,769 CCG

to CUG; 6,886 nt; LY75-Segment). In some embodiments, an LY75 variant mRNA molecule is any of the mRNA molecule isoforms described above produced from the LY75 variant genomic nucleic acid molecule rs147820690 comprising a C>T variation at position chr2:159878663

20 (GRCh38.p13; NC_000002.12:g.159878663C>T).

The nucleotide sequence of an LY75 reference cDNA molecule is set forth in SEQ ID

NO:25. The nucleotide sequence of another LY75 reference cDNA molecule is set forth in SEQ

ID NO:26. The nucleotide sequence of another LY75 reference cDNA molecule is set forth in

SEQ ID NO:27. The nucleotide sequence of another LY75 reference cDNA molecule is set forth

25 in SEQ ID NO:28. The nucleotide sequence of another LY75 reference cDNA molecule is set

forth in SEQ ID NO:29. The nucleotide sequence of another LY75 reference cDNA molecule is

set forth in SEQ ID NO:30. The nucleotide sequence of another LY75 reference cDNA molecule

is set forth in SEQ ID NO:31. The nucleotide sequence of another LY75 reference cDNA

molecule is set forth in SEQ ID NO:32. The nucleotide sequence of another LY75 reference

30 cDNA molecule is set forth in SEQ ID NO:33. The nucleotide sequence of another LY75

reference cDNA molecule is set forth in SEQ ID NO:34. The nucleotide sequence of another

LY75 reference cDNA molecule is set forth in SEQ ID NO:35.

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The nucleotide sequence of an LY75 variant cDNA molecule is set forth in SEQ ID NO:36. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:37. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:38. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:39. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:40. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:41. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:42. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:43. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:44. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:45. The nucleotide sequence of another LY75 variant cDNA molecule is set forth in SEQ ID NO:46. In some embodiments, an LY75 variant cDNA molecule is any cDNA molecule produced from any mRNA isoform molecule produced from the LY75 variant genomic nucleic acid molecule rs147820690 comprising a C>T variation at position chr2:159878663 (GRCh38.p13; NC_000002.12:g.159878663C>T).

The amino acid sequence of an LY75 reference polypeptide is set forth in SEQ ID NO:47 (Isoform 1; AAC17636.1) and is 1,722 amino acids in length. The amino acid sequence of another LY75 reference polypeptide is set forth in SEQ ID NO:48 (Isoform 2; NP_001185688.1) and is 1,873 amino acids in length. The amino acid sequence of another LY75 reference polypeptide is set forth in SEQ ID NO:49 (Isoform 3; NP_001185689.1) and is 1,817 amino acids in length. The amino acid sequence of another LY75 reference polypeptide is set forth in SEQ ID NO:50 (Isoform 4; BAD92152.1) and is 1,340 amino acids in length.

The amino acid sequence of an LY75 variant polypeptide is set forth in SEQ ID NO:51 (Isoform 1; AAC17636.1; Pro1,247Leu) and is 1,722 amino acids in length. The amino acid sequence of another LY75 variant polypeptide is set forth in SEQ ID NO:52 (Isoform 2; NP_001185688.1; Pro1,247Leu) and is 1,873 amino acids in length. The amino acid sequence of another LY75 variant polypeptide is set forth in SEQ ID NO:53 (Isoform 3; NP_001185689.1; Pro1,247Leu) and is 1,817 amino acids in length. The amino acid sequence of another LY75 variant polypeptide is set forth in SEQ ID NO:54 (Isoform 4; BAD92152.1; Pro865Leu) and is 1,340 amino acids in length. In some embodiments, an LY75 variant polypeptide is G525E produced from the LY75 variant genomic nucleic acid molecule rs147820690 comprising a C>T variation at position chr2:159878663 (GRCh38.p13; NC_000002.12:g.159878663C>T).

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The nucleotide sequence of a CD164 reference genomic nucleic acid molecule is set forth in SEQ ID NO:55 (ENSG00000135535.1 encompassing chr6:109,366,514-109,381,739 in the GRCh38/hg38 human genome assembly). The nucleotide sequence of a CD164 variant genomic nucleic acid molecule is set forth in SEQ ID NO:56 (rs3799840; T297A; 15,226 bp).

5 The nucleotide sequence of a CD164 reference mRNA molecule is set forth in SEQ ID NO:57 (NM_001346500; Isoform 1; 2,992 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:58 (ENST00000413644.6; Isoform 2; 2,414 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:59 (NM_006016.6; Isoform; 3 3,020 nt). The nucleotide sequence of another CD164
10 reference mRNA molecule is set forth in SEQ ID NO:60 (ENST00000275080.11; Isoform 4; 2,954 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:61 (ENST00000324953.9; Isoform 5; 2,936 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:62 (ENST00000512821.5; Isoform 6; 964 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID
15 NO:63 (NM_001142403.3; Isoform 7; 2,424 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:64 (NM_001142402.3; Isoform 8; 2,963 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:65 (NM_001142401.3; Isoform 9; 2,981 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:66 (D14043.1; Isoform 10; 2,427 nt). The
20 nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:67 (AF299341.1; Isoform 11; 2,929 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:68 (AF299342.1; Isoform 12; 2,950 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:69 (AF299343.1; Isoform 13; 2,968 nt). The nucleotide sequence of another CD164 reference
25 mRNA molecule is set forth in SEQ ID NO:70 (BC011522.3; Isoform 14; 3,010 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:71 (AK301692.1; Isoform 15; 1,294 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:72 (AK303525.1; Isoform 16; 1,386 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:73
30 (AK315908.1; Isoform 17; 1,386 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:74 (AF106518.1; Isoform; 18 537 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:75

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(AF263279.1; Isoform 19; 594 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:76 (FJ200494.1; Isoform 20; 590 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:77 (AK312357.1; Isoform 21; 683 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:78 (ENST00000368961.6; Isoform 22; 3,106 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:79 (ENST00000310786.5; Isoform 23; 2,993 nt). The nucleotide sequence of another CD164 reference mRNA molecule is set forth in SEQ ID NO:80 (ENST00000504373.1; Isoform 24; 1,402 nt).

The nucleotide sequence of a CD164 reference cDNA molecule is set forth in SEQ ID NO:81. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:82. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:83. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:84. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:85. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:86. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:87. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:88. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:89. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:90. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:91. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:92. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:93. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:94. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:95. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:96. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:97. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:98. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:99. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:100. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:101. The nucleotide sequence of another CD164 reference cDNA

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molecule is set forth in SEQ ID NO:102. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:103. The nucleotide sequence of another CD164 reference cDNA molecule is set forth in SEQ ID NO:104.

The amino acid sequence of a CD164 reference polypeptide is set forth in SEQ ID NO:105 (Isoform 1), and is 163 amino acids in length. The amino acid sequence of another CD164 reference polypeptide is set forth in SEQ ID NO:106 (NP_001135875.1; Isoform 2), and is 189 amino acids in length. The amino acid sequence of another CD164 reference polypeptide is set forth in SEQ ID NO:107 (AAG53906.1; Isoform 3), and is 197 amino acids in length. The amino acid sequence of another CD164 reference polypeptide is set forth in SEQ ID NO:108 (NP_001135873.1; Isoform 4), and is 184 amino acids in length. The amino acid sequence of another CD164 reference polypeptide is set forth in SEQ ID NO:109 (NP_001135874.1; Isoform 5), and is 178 amino acids in length. The amino acid sequence of another CD164 reference polypeptide is set forth in SEQ ID NO:110 (Isoform 6), and is 157 amino acids in length. The amino acid sequence of another CD164 reference polypeptide is set forth in SEQ ID NO:111 (BAG63164.1; Isoform 7), and is 156 amino acids in length. The amino acid sequence of another CD164 reference polypeptide is set forth in SEQ ID NO:112 (ACO54891.1; Isoform 8), and is 147 amino acids in length.

The nucleotide sequence of a PARP1 reference genomic nucleic acid molecule is set forth in SEQ ID NO:113 (ENSG00000143799.14 encompassing chr1:226,360,691-226,408,093 in the GRCh38/hg38 human genome assembly).

The nucleotide sequence of a PARP1 reference mRNA molecule is set forth in SEQ ID NO:114 (ENST00000366794.10; Isoform 1; 3,978 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:115 (ENST00000677203.1; Isoform 2; 3,850 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:116 (J03473.1; Isoform 3; 3,795 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:117 (BC037545; Isoform 4; 3,677 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:118 (M18112.1; Isoform 5; 3,640 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:119 (M32721.1; Isoform 6; 3,660 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:120 (AK303340.1; Isoform 7; 3,371 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:121 (M17081.1; Isoform 8; 1,771 nt). The nucleotide

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sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:122 (AK312339.1; Isoform 9; 3,132 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:123 (BC018620.1; Isoform 10; 827 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:124 (BC014206; Isoform 11; 902 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:125 (ENST00000366792.3; Isoform 12; 553 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:126 (ENST00000629232.1; Isoform 13; 477 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:127 (ENST00000366790.3; Isoform 14; 570 nt). The nucleotide sequence of another PARP1 reference mRNA molecule is set forth in SEQ ID NO:128 (ENST00000366794.6; Isoform 15; 3,958 nt).

The nucleotide sequence of a PARP1 reference cDNA molecule is set forth in SEQ ID NO:129. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:130. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:131. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:132. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:133. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:134. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:135. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:136. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:137. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:138. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:139. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:140. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:141. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:142. The nucleotide sequence of another PARP1 reference cDNA molecule is set forth in SEQ ID NO:143.

The amino acid sequence of a PARP1 reference polypeptide is set forth in SEQ ID NO:144 (AAB59447.1; Isoform 1), and is 1,014 amino acids in length. The amino acid sequence of another PARP1 reference polypeptide is set forth in SEQ ID NO:145 (Isoform 2), and is 971 amino acids in length. The amino acid sequence of another PARP1 reference polypeptide is set

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forth in SEQ ID NO:146 (BAG64403.1; Isoform 3), and is 993 amino acids in length. The amino acid sequence of another PARP1 reference polypeptide is set forth in SEQ ID NO:147 (AAA51599.1; Isoform 4), and is 574 amino acids in length. The amino acid sequence of another PARP1 reference polypeptide is set forth in SEQ ID NO:148 (AAH18620.1; Isoform 5), and is 232 amino acids in length. The amino acid sequence of another PARP1 reference polypeptide is set forth in SEQ ID NO:149 (AAH14206.1; Isoform 6), and is 250 amino acids in length. The amino acid sequence of another PARP1 reference polypeptide is set forth in SEQ ID NO:150 (Isoform 7), and is 108 amino acids in length. The amino acid sequence of another PARP1 reference polypeptide is set forth in SEQ ID NO:151 (Isoform 8), and is 155 amino acids in length.

The nucleotide sequence of a DNMT3A reference genomic nucleic acid molecule is set forth in SEQ ID NO:212 (ENSG00000119772.17 encompassing chr2:25,227,855-25,342,590 in the GRCh38/hg38 human genome assembly).

The nucleotide sequence of a DNMT3A reference mRNA molecule is set forth in SEQ ID NO:213 (ENST00000264709.7; Isoform 1; 9,501 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:214 (ENST00000321117.10; Isoform 2; 9,421 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:215 (ENST00000406659.3; Isoform 3; 1,775 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:216 (ENST00000380746.8; Isoform 4; 3,589 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:217 (ENST00000402667.1; Isoform 5; 2,300 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:218 (NM_175629.2; Isoform 6; 4,395 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:219 (NM_001320892.2; Isoform 7; 1,714 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:220 (NM_175630.1; Isoform 8; 1,808 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:221 (NM_001320893.1; Isoform 9; 3,638 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:222 (NM_153759.3; Isoform 10; 3,608 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:223 (NM_001375819.1; Isoform 11; 3,473 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:224 (BC043617.1; Isoform 12; 4,294 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:225 (AF331856.1; Isoform 13; 4,258 nt). The

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nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:226 (AB208833.1; Isoform 14; 4,476 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:227 (BC018214.1; Isoform 15; 1,758 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:228 (AF480163.1; Isoform 16; 2,371 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:229 (BC023612.2; Isoform 17; 1,113 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:230 (AF067972.2; Isoform 18; 3,005 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:231 (BC051864.1; Isoform 19; 943 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:232 (ENST00000321117.9; Isoform 20; 4,279 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:233 (ENST00000380756.4; Isoform 21; 4,477 nt). The nucleotide sequence of another DNMT3A reference mRNA molecule is set forth in SEQ ID NO:234 (ENST00000683760.1; Isoform 22; 3,585 nt).

The nucleotide sequence of a DNMT3A reference cDNA molecule is set forth in SEQ ID NO:235. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:236. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:237. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:238. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:239. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:240. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:241. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:242. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:243. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:244. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:245. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:246. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:247. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:248. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:249. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:250. The nucleotide sequence of another DNMT3A

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reference cDNA molecule is set forth in SEQ ID NO:251. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:252. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:253. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:254. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:255. The nucleotide sequence of another DNMT3A reference cDNA molecule is set forth in SEQ ID NO:256.

The amino acid sequence of a DNMT3A reference polypeptide is set forth in SEQ ID NO:257 (NP_783328.1; Isoform 1), and is 912 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:258 (NP_001307821.1; Isoform 2), and is 166 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:259 (NP_715640.2; Isoform 3), and is 723 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:260 (NP_001362748.1; Isoform 4), and is 689 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:261 (NP_001307822.1; Isoform 5), and is 760 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:262 (AAL57039.1; Isoform 6), and is 909 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:263 (BAD92070.1; Isoform 7), and is 811 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:264 (AAH18214.1; Isoform 8), and is 285 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:265 (AAH23612.1; Isoform 9), and is 351 amino acids in length. The amino acid sequence of another DNMT3A reference polypeptide is set forth in SEQ ID NO:266 (AAH23612.1; Isoform 10), and is 781 amino acids in length.

The nucleotide sequence of an ASXL1 reference genomic nucleic acid molecule is set forth in SEQ ID NO:267 (ENSG00000171456.20 encompassing chr20:32,358,330-32,439,260 in the GRCh38/hg38 human genome assembly).

The nucleotide sequence of an ASXL1 reference mRNA molecule is set forth in SEQ ID NO:268 (ENST00000651418.1; Isoform 1; 3,146 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:269 (ENST00000375687.10; Isoform 2; 7,052 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in

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SEQ ID NO:270 (ENST00000542461.5; Isoform 3; 1,068 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:271 (ENST00000613218.4; Isoform 4; 7,038 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:272 (ENST00000646367.1; Isoform 5; 1,065 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:273 (ENST00000620121.4; Isoform 6; 5,374 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:274 (ENST00000646985.1; Isoform 7; 6,666 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:275 (ENST00000497249.6; Isoform 8; 495 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:276 (ENST00000375689.5; Isoform 9; 812 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:277 (ENST00000306058.9; Isoform 10; 6,591 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:278 (NM_001164603.1; Isoform 11; 1,084 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:279 (BC100280.1; Isoform 12; 1,078 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:280 (BC064984.1; Isoform 13; 1,009 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:281 (AJ438952.2; Isoform 14; 6,864 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:282 (AK122923.1; Isoform 15; 4,685 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:283 (AB023195.2; Isoform 16; 6,088 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:284 (AL117518.1; Isoform 17; 4,055 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:285 (ENST00000375687.5; Isoform 18; 7,031 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:286 (ENST00000497249.2; Isoform 19; 296 nt). The nucleotide sequence of another ASXL1 reference mRNA molecule is set forth in SEQ ID NO:287 (ENST00000555343.2; Isoform 20; 1,034 nt).

The nucleotide sequence of an ASXL1 reference cDNA molecule is set forth in SEQ ID NO:288. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:289. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:290. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:291. The nucleotide sequence of another ASXL1 reference cDNA molecule is

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set forth in SEQ ID NO:292. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:293. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:294. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:295. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:296. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:297. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:298. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:299. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:300. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:301. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:302. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:303. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:304. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:305. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:306. The nucleotide sequence of another ASXL1 reference cDNA molecule is set forth in SEQ ID NO:307.

The amino acid sequence of an ASXL1 reference polypeptide is set forth in SEQ ID NO:308 (Isoform 1), and is 625 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:309 (CAD27708.1; Isoform 2), and is 1,541 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:310 (NP_001158075.1; Isoform 3), and is 85 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:311 (Isoform 4), and is 1,480 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:312 (Isoform 5), and is 75 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:313 (Isoform 6), and is 81 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:314 (Isoform 7), and is 1,536 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:315 (AAH64984.1; Isoform 8), and is 84 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:316 (BAG53800.1; Isoform 9), and is 1,462 amino acids in length. The amino acid sequence of another ASXL1 reference

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polypeptide is set forth in SEQ ID NO:317 (BAA76822.2; Isoform 10), and is 1,368 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:318 (Isoform 11), and is 1,341 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:319 (Isoform 12), and is 60 amino acids in length. The amino acid sequence of another ASXL1 reference polypeptide is set forth in SEQ ID NO:320 (Isoform 13), and is 57 amino acids in length.

The genomic nucleic acid molecules, mRNA molecules, and cDNA molecules can be from any organism. For example, the genomic nucleic acid molecules, mRNA molecules, and cDNA molecules can be human or an ortholog from another organism, such as a non-human mammal, a rodent, a mouse, or a rat. It is understood that gene sequences within a population can vary due to polymorphisms such as single-nucleotide polymorphisms. The examples provided herein are only exemplary sequences. Other sequences are also possible.

Also provided herein are functional polynucleotides that can interact with the disclosed nucleic acid molecules. Examples of functional polynucleotides include, but are not limited to, antisense molecules, aptamers, ribozymes, triplex forming molecules, and external guide sequences. The functional polynucleotides can act as effectors, inhibitors, modulators, and stimulators of a specific activity possessed by a target molecule, or the functional polynucleotides can possess a *de novo* activity independent of any other molecules.

The isolated nucleic acid molecules disclosed herein can comprise RNA, DNA, or both RNA and DNA. The isolated nucleic acid molecules can also be linked or fused to a heterologous nucleic acid sequence, such as in a vector, or a heterologous label. For example, the isolated nucleic acid molecules disclosed herein can be within a vector or as an exogenous donor sequence comprising the isolated nucleic acid molecule and a heterologous nucleic acid sequence. The isolated nucleic acid molecules can also be linked or fused to a heterologous label. The label can be directly detectable (such as, for example, fluorophore) or indirectly detectable (such as, for example, hapten, enzyme, or fluorophore quencher). Such labels can be detectable by spectroscopic, photochemical, biochemical, immunochemical, or chemical means. Such labels include, for example, radiolabels, pigments, dyes, chromogens, spin labels, and fluorescent labels. The label can also be, for example, a chemiluminescent substance; a metal-containing substance; or an enzyme, where there occurs an enzyme-dependent secondary generation of signal. The term "label" can also refer to a "tag" or hapten that can bind selectively to a conjugated molecule such that the conjugated molecule, when added

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subsequently along with a substrate, is used to generate a detectable signal. For example, biotin can be used as a tag along with an avidin or streptavidin conjugate of horseradish peroxidase (HRP) to bind to the tag, and examined using a calorimetric substrate (such as, for example, tetramethylbenzidine (TMB)) or a fluorogenic substrate to detect the presence of HRP. Exemplary labels that can be used as tags to facilitate purification include, but are not limited to, myc, HA, FLAG or 3XFLAG, 6Xhis or polyhistidine, glutathione-S-transferase (GST), maltose binding protein, an epitope tag, or the Fc portion of immunoglobulin. Numerous labels include, for example, particles, fluorophores, haptens, enzymes and their calorimetric, fluorogenic and chemiluminescent substrates and other labels.

Percent identity (or percent complementarity) between particular stretches of nucleotide sequences within nucleic acid molecules or amino acid sequences within polypeptides can be determined routinely using BLAST programs (basic local alignment search tools) and PowerBLAST programs (Altschul *et al.*, J. Mol. Biol., 1990, 215, 403-410; Zhang and Madden, Genome Res., 1997, 7, 649-656) or by using the Gap program (Wisconsin Sequence Analysis Package, Version 8 for Unix, Genetics Computer Group, University Research Park, Madison Wis.), using default settings, which uses the algorithm of Smith and Waterman (Adv. Appl. Math., 1981, 2, 482-489). Herein, if reference is made to percent sequence identity, the higher percentages of sequence identity are preferred over the lower ones.

As used herein, the phrase "corresponding to" or grammatical variations thereof when used in the context of the numbering of a particular nucleotide or nucleotide sequence or position refers to the numbering of a specified reference sequence when the particular nucleotide or nucleotide sequence is compared to a reference sequence (such as, for example, SEQ ID NO:1, SEQ ID NO:55, or SEQ ID NO:113). In other words, the residue (such as, for example, nucleotide or amino acid) number or residue (such as, for example, nucleotide or amino acid) position of a particular polymer is designated with respect to the reference sequence rather than by the actual numerical position of the residue within the particular nucleotide or nucleotide sequence. For example, a particular nucleotide sequence can be aligned to a reference sequence by introducing gaps to optimize residue matches between the two sequences. In these cases, although the gaps are present, the numbering of the residue in the particular nucleotide or nucleotide sequence is made with respect to the reference sequence to which it has been aligned.

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The nucleotide and amino acid sequences listed in the accompanying sequence listing are shown using standard letter abbreviations for nucleotide bases, and three-letter code for amino acids. The nucleotide sequences follow the standard convention of beginning at the 5' end of the sequence and proceeding forward (i.e., from left to right in each line) to the 3' end.

5 Only one strand of each nucleotide sequence is shown, but the complementary strand is understood to be included by any reference to the displayed strand. The amino acid sequence follows the standard convention of beginning at the amino terminus of the sequence and proceeding forward (i.e., from left to right in each line) to the carboxy terminus.

The present disclosure also provides methods of stratifying a suitable lung cancer
10 patient for treatment with a PARP1 inhibitor. The methods comprise determining whether the patient carries a DNMT3A R882H somatic mutation or TET2 somatic mutation deficiency. The patient with the DNMT3A R882H somatic mutation or TET2 somatic mutation deficiency can be excluded from a PARP1 inhibitor treatment regimen.

The present disclosure also provides therapeutic agents that prevent or reduce CHIP
15 for use in the prevention and/or reduction of CHIP in a subject having: an LY75 variant genomic nucleic acid molecule, a CD164 variant genomic nucleic acid molecule, and/or a PARP1 variant genomic nucleic acid molecule; an LY75 variant mRNA molecule, a CD164 variant mRNA molecule, and/or a PARP1 variant mRNA molecule; or an LY75 variant cDNA molecule, a CD164 variant cDNA molecule, and/or a PARP1 variant cDNA molecule. Any of the therapeutic agents
20 that prevent or reduce CHIP described herein can be used in these methods.

The present disclosure also provides uses of therapeutic agents that prevent or reduce CHIP for use in the preparation of a medicament for prevention or reduction of CHIP in a subject having: an LY75 variant genomic nucleic acid molecule, a CD164 variant genomic nucleic acid molecule, and/or a PARP1 variant genomic nucleic acid molecule; an LY75 variant mRNA
25 molecule, a CD164 variant mRNA molecule, and/or a PARP1 variant mRNA molecule; or an LY75 variant cDNA molecule, a CD164 variant cDNA molecule, and/or a PARP1 variant cDNA molecule. Any of the therapeutic agents that prevent or reduce CHIP described herein can be used in these methods.

The present disclosure also provides an LY75 inhibitor for use in the prevention or
30 reduction of CHIP in a subject having: an LY75 variant genomic nucleic acid, an LY75 variant mRNA molecule, or an LY75 variant cDNA molecule. Any of the LY75 inhibitors described herein can be used in these methods.

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The present disclosure also provides a CD164 inhibitor for use in the prevention or reduction of CHIP in a subject having: a CD164 variant genomic nucleic acid molecule, a CD164 variant mRNA molecule, or a CD164 variant cDNA molecule. Any of the CD164 inhibitors described herein can be used in these methods.

5 The present disclosure also provides a PARP1 inhibitor for use in the prevention or reduction of CHIP in a subject having: a PARP1 variant genomic nucleic acid molecule, a PARP1 variant mRNA molecule, or a PARP1 variant cDNA molecule. Any of the PARP1 inhibitors described herein can be used in these methods.

10 The present disclosure also provides an LY75 inhibitor for use in the preparation of a medicament for the prevention or reduction of CHIP in a subject having: an LY75 variant genomic nucleic acid molecule, an LY75 variant mRNA molecule, or an LY75 variant cDNA molecule. Any of the LY75 inhibitors described herein can be used in these methods.

15 The present disclosure also provides a CD164 inhibitor for use in the preparation of a medicament for the prevention or reduction of CHIP in a subject having: a CD164 variant genomic nucleic acid molecule, a CD164 variant mRNA molecule, or a CD164 variant cDNA molecule. Any of the CD164 inhibitors described herein can be used in these methods.

20 The present disclosure also provides a PARP1 inhibitor for use in preparation of a medicament for the prevention or reduction of CHIP in a subject having: a PARP1 variant genomic nucleic acid molecule, a PARP1 variant mRNA molecule, or a PARP1 variant cDNA molecule. Any of the PARP1 inhibitors described herein can be used in these methods.

25 All patent documents, websites, other publications, accession numbers and the like cited above or below are incorporated by reference in their entirety for all purposes to the same extent as if each individual item were specifically and individually indicated to be so incorporated by reference. If different versions of a sequence are associated with an accession number at different times, the version associated with the accession number at the effective filing date of this application is meant. The effective filing date means the earlier of the actual filing date or filing date of a priority application referring to the accession number if applicable. Likewise, if different versions of a publication, website or the like are published at different times, the version most recently published at the effective filing date of the application is
30 meant unless otherwise indicated. Any feature, step, element, embodiment, or aspect of the present disclosure can be used in combination with any other feature, step, element, embodiment, or aspect unless specifically indicated otherwise. Although the present disclosure

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has been described in some detail by way of illustration and example for purposes of clarity and understanding, it will be apparent that certain changes and modifications may be practiced within the scope of the appended claims.

The following examples are provided to describe the embodiments in greater detail.

- 5 They are intended to illustrate, not to limit, the claimed embodiments. The following examples provide those of ordinary skill in the art with a disclosure and description of how the compounds, compositions, articles, devices and/or methods described herein are made and evaluated, and are intended to be purely exemplary and are not intended to limit the scope of any claims. Efforts have been made to ensure accuracy with respect to numbers (such as, for
10 example, amounts, temperature, etc.), but some errors and deviations may be accounted for. Unless indicated otherwise, parts are parts by weight, temperature is in °C or is at ambient temperature, and pressure is at or near atmospheric.

Examples

15 **Example 1: Common Variants in LY75/CD302/LY75-CD302 Locus Associate with CHIP**

Several common variants in LY75/CD302/LY75-CD302 locus associate with CHIP, including LY75 missense rs78446341 (see, Table 4).

Table 4: LY75/CD302/LY75-CD302 locus associations with CHIP

Variant	rsID	P-value	Effect	MAF	CADD	varEffect	Lof
2:159868900:C:A	rs56186131	2.83E-12	1.058	0.4569	0.371	intronic	No
2:159861821:C:G	rs72955755	3.13E-12	1.058	0.4562	0.676	intronic	No
2:159870033:T:G	rs17231342	3.94E-12	1.058	0.4560	4.066	intronic	No
2:159834145:G:A	rs78446341	2.48E-08	0.8541	0.0227	24.5	missense	No

20

Table 4 (cont.): LY75/CD302/LY75-CD302 locus associations with CHIP

Variant	rsID	eQTL	min_eQTL LPval	is Path	In HGM D	In GWAS ctlg	nGWA Sctlg Traits	minGWA SctlgPval
2:159868900:C:A	rs56186131	FALSE	NA	FALSE	FALSE	FALSE	0	NA
2:159861821:C:G	rs72955755	TRUE	2.51E-14	FALSE	FALSE	FALSE	0	NA
2:159870033:T:G	rs17231342	TRUE	9.17E-14	FALSE	FALSE	FALSE	0	NA
2:159834145:G:A	rs78446341	FALSE	NA	FALSE	TRUE	TRUE	1	4.00E-29

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Common LY75-CD302 locus variants that are in eQTLs for nearby genes are shown in Table 5.

Table 5: Common LY75-CD302 locus variants are in eQTLs for nearby genes

uid	geneName	Pval	Dataset
2:159861821:C:G	AC009961.1	2.51e-14	RegnGHS_Liver
2:159870033:T:G	MARCH7	3.403e-13	Westra_Whole_Blood
2:159870033:T:G	MARCH7	9.1657e-14	Vosa_Whole_Blood

5

Table 5 (cont.): Common LY75-CD302 locus variants are in eQTLs for nearby genes

uid	Cis_Trans	Effect_Allele	Ref_Allele	Effects
2:159861821:C:G	CIS	G	C	0.214
2:159870033:T:G	CIS	G	T	-7.2774
2:159870033:T:G	CIS	G	T	-7.4524

Finemapping the *LY75-CD302* locus identified rs78446341 as highly likely (~80%) to be driving the association signal at this locus (data not shown). In addition, all LY75 burden masks suggest a reduced CHIP risk (see, Figure 1). Moreover, the effect of the ly75 locus varies by CHIP gene mutation carrier (see, Figure 2).

LY75 missense p.Pro1247Leu significantly associated with platelets, neutrophils, and lymphocytes in UKB 500K (see, Table 6; Variant = 2:159834145:G:A; HGVS = p.Pro1247Leu).

15

Table 6: LY75 missense p.Pro1247Leu significantly associates with platelets, neutrophils, and lymphocytes in UKB 500K

Phenotype	Effect (95% CI)	P-value	Cases RR RA AA	Controls RR RA AA	AAF
Platelet crit	↑ 0.115 (0.103, 0.128)	8.3E-76	NA	399599 19216 245	0.024
Neutrophils	↑ 0.047 (0.034, 0.061)	2.7E-12	NA	398255 19282 242	0.024
Lymphocytes	↓ -0.046 (-0.060, -0.033)	4.5E-12	NA	398260 19282 242	0.024

Additional rare pLoFs and rare missense variants in LY75 also associated with reduced odds of CHIP (see, Table 7, Phenotype = CHIP; and Figure 3).

20

Table 7: Additional rare pLoFs and rare missense variants in LY75 also associate with reduced odds of CHIP

Mask	Variants in Mask	Effect (95% CI)	P-value	Cases RR RA AA	Controls RR RA AA	AAF

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M1.1	pLoFs, <1% AAF	0.78 (0.68, 0.89)	2.6E-4	34385 215 0	499850 4037 2	0.004
M1.01	pLoFs, <0.1% AAF	0.75 (0.61, 0.92)	6.5E-3	34504 96 0	502106 1783 0	0.002
M3.1	pLoFs & missense, <1% AAF	0.78 (0.68, 0.90)	4.3E-4	34283 218 0	499841 4046 2	0.004
M3.01	pLoFs & missense, <0.1% AAF	0.77 (0.63, 0.94)	0.012	34501 99 0	502096 1792 0	0.002

Example 2: Common Variants in *CD164* Locus Associate with CHIP

Multiple common variant signals in *CD164* locus associate with CHIP, although fine-mapping does not point to a causal gene (data not shown). In addition, eQTL data supports variant effects on *CD164* expression (see, Table 8).

Table 8: eQTL data variant effects on *CD164* expression

Uid	geneName	Pval	Dataset
6:109287294:C:T	CD164	5.56e-14	Fairfax_Monocytes_LPS24
6:109326927:T:G	CD164	5.58008e-19	GTeXv*_Whole_Blood
6:109381443:A:T	CD164	4.037e-29	Chen_Neutrophils

Table 8 (cont.): eQTL data variant effects on *CD164* expression

uid	Cis_Trans	Effect_Allele	Ref_Allele	Effects
6:109287294:C:T	CIS	NA	NA	NA
6:109326927:T:G	CIS	G	T	0.123912
6:109381443:A:T	CIS	T	A	-1.117

Example 3: Common Variants in *PARP1* Locus Associate with CHIP

A *PARP1* 1:226367601:A:C missense variant was in perfect LD with index SNP and showed a protective association (see, Figure 4). Other CHIP-associated variants were significant eQTLs for *PARP1* (see, Figure 5).

Example 4: Somatic Mutations in *DNMT3A* and/or *ASXL1* Associate with Increased Risk of Developing Lung Cancer

Exome sequencing data from the UKB Exome Sequencing Consortium was used to identify CHIP somatic mutation carriers across 454,787 UKB participants. This was complemented by generating an additional CHIP callset across 133,370 individuals from the DiscoverEHR Cohort. These represent the largest CHIP callsets to date, and were used to

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conduct genetic association analyses of CHIP across 29,669 CHIP mutation carriers from UKB, and to perform replication in 14,766 CHIP mutation carriers from the DiscoverEHR cohort. 27 loci associated with CHIP were identified in UKB at genome-wide significance, which was replicated in DiscoverEHR. Additionally, phenotypic associations for both CHIP somatic
5 mutation carriers and germline CHIP risk loci were investigated across 35,000 traits from the UK Biobank.

The analysis described herein only includes CHIP carriers with the largest expansion of blood cells with the CHIP mutations (estimated to have $\geq \sim 20\%$ of their blood cells with the CHIP mutations. It was determined whether CHIP carriers were at an elevated risk of
10 developing solid tumors (see Figure 6), and it was found that high variant allele fraction (VAF) carriers are at significantly elevated risk of developing lung cancer ($HR = 1.62 (1.41-1.87)$, $p = 1.36 \times 10^{-11}$), and modestly increased risk of developing prostate ($HR = 1.17 (1.04-1.31)$, $p = 7.1 \times 10^{-3}$) and breast cancers ($HR = 1.15 (1.002-1.322)$, $p = 0.047$). No increased risk for the development of colon cancer was found ($HR = 0.96 (0.79-1.16)$, $p = 0.66$). Models estimating
15 event risk on the basis of CHIP mutational subtype (e.g., carriers must have DNMT3A mutations) suggest that these associations with prostate and breast cancer are driven primarily by DNMT3A mutations.

Given the strong associations between CHIP and blood cancer and lung cancer, and the associations between smoking and both CHIP and lung cancer, additional analyses stratified by
20 smoking status were performed to test whether these associations were driven by smoking and merely marked by CHIP mutations. High VAF CHIP carriers are at an elevated risk of developing blood cancers in smokers ($2.37 (1.86-3.03)$, $p = 5.24 \times 10^{-12}$) and non-smokers ($2.05 (1.68-2.49)$, $P = 7.47 \times 10^{-13}$), and this pattern was similar among all CHIP carriers and across associations with CHIP mutation subtype ASXL1-CHIP. Lung cancer risk among all CHIP carriers
25 is the same among smokers ($HR = 1.45 (1.28-1.63)$, $p = 1.10 \times 10^{-9}$) and non-smokers ($HR = 1.45 (1.18-1.80)$, $P = 5.46 \times 10^{-4}$), and is actually higher in non-smokers among CHIP carriers with $VAF \geq 10\%$ ($HR_{smokers} = 1.56 (1.33-1.84)$, $p = 7.32 \times 10^{-8}$ compared to $HR_{non-smokers} = 1.93 (1.47-2.54)$, $P = 2.86 \times 10^{-6}$). When sub-setting to DNMT3A-CHIP and ASXL1-CHIP, the patterns were similar. Overall, the models suggest that CHIP mutation carriers are at an elevated risk of
30 blood cancer and lung cancer independent of smoking, but that CHIP is likely also marking additional blood cancer risk that results from smoking.

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In summary, in longitudinal analyses, it was found that individuals who carry CHIP somatic mutations in either DNMT3A or ASXL1 have an increased risk of lung cancer in both smokers and non-smokers, which indicates that CHIP is an independent risk factor for malignant neoplasms outside of the hemopoietic lineage.

5

Example 5: Effects of PARPi on Cells Harboring CHIP Gene Mutation

To determine the effect, if any, that PARP1 inhibitors have on cells *in vitro* that carry CHIP gene mutations, variants/mutations were engineered into cell lines using CRISPR-Cas9 and specifically optimized sgRNA target sequence as well donor sequence for the DNMT3A Knockin.

10 Functional modeling of identified CHIP-GWAS locus was performed. Selected PARP1 locus from CHIP-GWAS analysis showed significant association of DNMT3A to protective PARP1 germline variant (data not shown). Figure 7, Panels A and B show systematic tracking of HDR and INDEL percentages in PARPi treated cells. Cells were treated with PARPi for 8 days and then subjected to Sanger sequencing across each locus to determine population dynamics. Figure 7, Panel A shows RPE-1 hTERT cells harboring CRISPR mediated knock-in of DNMT3A-R882H allele. This shows that there is no effect of PARP1 inhibitor treatment on cells carrying the DNMT3A variant allele and it remains unchanged in the population as compared to DMSO (control/vehicle) alone. Thus, PARP1 inhibitors had no effect on cells carrying the DNMT3A-R882H variant. Referring to Figure 7, Panel A, the percentage represents the percent of sequences that derive from that represented allele in the legend, with all three legend items adding up to roughly 100% for each of the samples. Thus, the wild-type allele (blue group) for the DMSO group has an average of ~16% (16% of the sequences in the population of cells are wild-type). The R882H allele (black group) in DMSO has ~24% (24% of the sequences derived from the population of cells have the R882H mutation). The INDEL (red group) in DMSO has ~60% (60% of sequences in the population carry INDEL (nucleotide insertion/deletion) alleles). The total for wild-type, R882H, and INDEL for the DMSO group is ~100% of the sequences analyzed.

20 Figure 7, Panel B shows CRISPR-mediated TET2-deficiency in HEK293T cells. Cells carrying potential loss of function alleles (INDELs/KO alleles) are resistant to PARP1 inhibitor treatment -- in olaparib and talazoparib conditions, the percentage of INDELs (mutant alleles) increased compared to DMSO (control/vehicle). This means that cells carrying TET2 KO/INDEL alleles survive better than wild-type alleles in response to treatment with PARP1 inhibitors.

30

Thus, in this case, it may be advisable to avoid PARP1 inhibitor treatment in order to not increase clonal selection for those TET2 mutant/variant alleles that cause deficiency.

Example 6: Jointly Fine-Mapping Common and Rare Variants at the LY75 Locus

DNMT3A, which was the most commonly mutated gene in the overall CHIP phenotype, had the largest number of significantly associated loci ($n = 23$), most of which overlapped with the overall CHIP association signals. At a locus on chromosome 2, rs78446341 (P1247L in LY75) was associated with reduced risk of DNMT3A-CHIP ($OR = 0.78$ (0.72-0.84), $P = 3.70 \times 10^{-10}$), and was prioritized by fine-mapping. LY75 features lymphocyte specific expression, and is thought to be involved in antigen presentation and lymphocyte proliferation. A second rare (AAF=0.002) missense variant (rs147820690-T, G525E) was also identified that associated with reduced risk of DNMT3A-CHIP at close to genome-wide significance ($OR = 0.48$ (0.36-0.63), $P = 1.15 \times 10^{-7}$). This variant was predicted as likely to be damaging (CADD = 23.6) and remains associated ($OR = 0.63$ (0.51-0.77), $P = 4.80 \times 10^{-6}$) when conditioning on common variant signal in this locus (i.e., this rare variant signal is independent of the common variant signal in this locus). This variant was also prioritized by fine-mapping. Finally, these signals in PARP1 and LY75 replicated in GHS (see, Figure 8).

The more common LY75 missense variant (rs78446341-A, P1247L) is located in the extracellular domain of lymphocytic antigen 75, which is also known as DEC-205/CD205, and plays a role in antigenic capture, processing, and presentation. The rarer LY75 missense variant (rs147820690-T, G525E) is located in a C-type lectin domain and reported to interact directly with this receptor's ligand. The protective associations with this variant that were identified HEREIN appear to be most pronounced for DNMT3A-CHIP and mLOY and highlight LY75 as a therapeutic target for the antagonization of CH in general.

To further evaluate whether the rare variant association at the LY75 locus (rs147820690-T) was independent of other common and rare variant signals, joint fine-mapping (with FINEMAP) was performed on common and rare variants at this locus while including rarer variants then used in our genome-wide fine-mapping. In contrast to the genome-wide fine-mapping described above, this fine-mapping sensitivity analysis was done only in the UKB, was focused on the LY75 locus, and included all variants in the dataset. That is, the fine-mapping analysis was run as described above, but with a $MAF > 0.0000000001$. While FINEMAP suggests 3 credible sets are most parsimonious at this locus (posterior probability = 0.8), which is

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consistent with the results we report when performing genome-wide fine-mapping, the fourth credible set (posterior probability = 0.11) identifies rs147820690-T as the top signal (PIP = 0.133) among 9,417 variants in the 95% credible set. This fine-mapping approach also prioritizes rs78446341-A (CPIP = 0.92, CS = 2). Furthermore, the median pairwise LD between
5 SNPs in this fourth credible set is very low (6.7×10^{-4} , compared with 0.995, 0.962, and 0.831 for the first three credible sets, respectively). Therefore, these fine-mapping results provide additional support for both LY75 missense variants, as well as the fact that the rs147820690-T rare variant signal is not driven by the tagging of other rare variants.

10 Various modifications of the described subject matter, in addition to those described herein, will be apparent to those skilled in the art from the foregoing description. Such modifications are also intended to fall within the scope of the appended claims. Each reference (including, but not limited to, journal articles, U.S. and non-U.S. patents, patent application publications, international patent application publications, gene bank accession numbers, and
15 the like) cited in the present application is incorporated herein by reference in its entirety and for all purposes.

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What is Claimed is:

1. A method of preventing or reducing the development of clonal hematopoiesis of indeterminate potential (CHIP) in a subject, the methods comprising administering a Lymphocyte Antigen 75 (LY75) inhibitor, a Cluster of Differentiation 164 (CD164) inhibitor, or a
5 Poly(ADP-Ribose) Polymerase 1 (PARP1) inhibitor, or any combination thereof, to the subject.
2. The method according to claim 1, wherein the subject is at risk of developing a hematologic cancer, a myeloid neoplasia, a lymphoid neoplasia, an atherosclerotic cardiovascular disease, a coronary heart disease, a myocardial infarction, or severe calcified aortic valve stenosis.
- 10 3. The method according to claim 1 or claim 2, wherein the LY75 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to an LY75 nucleic acid molecule.
4. The method according to claim 3, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).
- 15 5. The method according to claim 1 or claim 2, wherein the LY75 inhibitor comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within an LY75 genomic nucleic acid molecule.
6. The method according to claim 5, wherein the Cas protein is Cas9 or Cpf1.
7. The method according to claim 5 or claim 6, wherein the LY75 gRNA recognition
20 sequence is located within SEQ ID NO:1.
8. The method according to claim 5 or claim 6, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.
9. The method according to any one of claims 5 to 8, wherein the gRNA comprises from
25 about 17 to about 23 nucleotides.
10. The method according to any one of claims 5 to 8, wherein the LY75 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs:152-171.
11. The method according to claim 1 or claim 2, wherein the CD164 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to a CD164 nucleic acid molecule.
- 30 12. The method according to claim 11, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).

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13. The method according to claim 1 or claim 2, wherein the CD164 inhibitor comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within a CD164 genomic nucleic acid molecule.
14. The method according to claim 13, wherein the Cas protein is Cas9 or Cpf1.
- 5 15. The method according to claim 13 or claim 14, wherein the CD164 gRNA recognition sequence is located within SEQ ID NO:55.
16. The method according to claim 13 or claim 14, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.
- 10 17. The method according to any one of claims 13 to 16, wherein the gRNA comprises from about 17 to about 23 nucleotides.
18. The method according to any one of claims 13 to 16, wherein the CD164 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs:172-191.
- 15 19. The method according to claim 1 or claim 2, wherein the PARP1 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to a PARP1 nucleic acid molecule.
20. The method according to claim 19, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).
- 20 21. The method according to claim 1 or claim 2, wherein the PARP1 inhibitor comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within a PARP1 genomic nucleic acid molecule.
22. The method according to claim 21, wherein the Cas protein is Cas9 or Cpf1.
23. The method according to claim 21 or claim 22, wherein the PARP1 gRNA recognition
25 sequence is located within SEQ ID NO:113.
24. The method according to claim 21 or claim 22, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.
25. The method according to any one of claims 21 to 24, wherein the gRNA comprises
30 from about 17 to about 23 nucleotides.

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26. The method according to any one of claims 21 to 24, wherein the PARP1 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs:192-211.

27. The method according to any one of claims 1 to 26, further comprising detecting the presence or absence of an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, or a PARP1 variant nucleic acid molecule, in a biological sample from the subject.

28. The method according to claim 27, further comprising administering a therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount to a subject wherein one or more of the LY75 variant nucleic acid molecule, CD164 variant nucleic acid molecule, or PARP1 variant nucleic acid molecule is absent from the biological sample.

29. The method according to claim 27, further comprising administering a therapeutic agent that prevents or reduces development of CHIP in a dosage amount that is the same as or less than a standard dosage amount to a subject that is heterozygous for one or more of the LY75 variant nucleic acid molecule, CD164 variant nucleic acid molecule, or PARP1 variant nucleic acid molecule.

30. The method according to any one of claims 27 to 29, wherein the LY75 variant nucleic acid molecule, CD164 variant nucleic acid molecule, or PARP1 variant nucleic acid molecule is a missense variant, splice-site variant, a stop-gain variant, a start-loss variant, a stop-loss variant, a frameshift variant, or an in-frame indel variant, or a variant that encodes a truncated predicted loss-of-function polypeptide.

31. The method according to claim 30, wherein the LY75 variant nucleic acid molecule encodes a truncated LY75 predicted loss-of-function polypeptide, the CD164 variant nucleic acid molecule encodes a truncated CD164 predicted loss-of-function polypeptide, or the PARP1 variant nucleic acid molecule encodes a truncated PARP1 predicted loss-of-function polypeptide.

32. A method of treating a subject with a therapeutic agent that prevents or reduces development of CHIP, wherein the subject has CHIP or is at risk of developing CHIP, the method comprising the steps of:

determining whether the subject has an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and/or a PARP1 variant nucleic acid molecule by:

obtaining or having obtained a biological sample from the subject;

and

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performing or having performed a sequence analysis on the biological sample to determine if the subject has a genotype comprising the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule; and

5 administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount to a subject that is LY75 reference, CD164 reference, and/or PARP1 reference; and/or administering an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject;

administering or continuing to administer the therapeutic agent that prevents or
10 reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is heterozygous for the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule; and/or administering an LY75 inhibitor, a CD164 inhibitor, or a PARP1 inhibitor, or any combination thereof, to the subject; or

15 administering or continuing to administer the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount to a subject that is homozygous for the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule;

wherein the presence of a genotype having the LY75 variant nucleic acid molecule, the
20 CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule indicates the subject has a decreased risk of developing CHIP.

33. The method according to claim 32, wherein the subject is at risk of developing a hematologic cancer, a myeloid neoplasia, a lymphoid neoplasia, an atherosclerotic cardiovascular disease, a coronary heart disease, a myocardial infarction, or severe calcified
25 aortic valve stenosis.

34. The method according to claim 32 or claim 33, wherein the subject is LY75 reference, and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount, and is administered the LY75 inhibitor.

30 35. The method according to claim 32 or claim 33, wherein the subject is CD164 reference, and the subject is administered or continued to be administered the therapeutic agent that

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prevents or reduces development of CHIP in a standard dosage amount, and is administered the CD164 inhibitor.

36. The method according to claim 32 or claim 33, wherein the subject is PARP1 reference, and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount, and is administered the PARP1 inhibitor.

37. The method according to claim 32 or claim 33, wherein the subject is heterozygous for the LY75 variant nucleic acid molecule, and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered the LY75 inhibitor.

38. The method according to claim 32 or claim 33, wherein the subject is heterozygous for the CD164 variant nucleic acid molecule, and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered the CD164 inhibitor.

39. The method according to claim 32 or claim 33, wherein the subject is heterozygous for the PARP1 variant nucleic acid molecule, and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is administered the PARP1 inhibitor.

40. The method according to any one of claims 32 to 39, wherein the LY75 variant nucleic acid molecule, CD164 variant nucleic acid molecule, or PARP1 variant nucleic acid molecule is a missense variant, splice-site variant, a stop-gain variant, a start-loss variant, a stop-loss variant, a frameshift variant, or an in-frame indel variant, or a variant that encodes a truncated predicted loss-of-function polypeptide.

41. The method according to claim 40, wherein the LY75 variant nucleic acid molecule encodes a truncated LY75 predicted loss-of-function polypeptide, the CD164 variant nucleic acid molecule encodes a truncated CD164 predicted loss-of-function polypeptide, or the PARP1 variant nucleic acid molecule encodes a truncated PARP1 predicted loss-of-function polypeptide.

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42. The method according to any one of claims 32 to 41, wherein the LY75 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to an LY75 nucleic acid molecule.

43. The method according to claim 42, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).

44. The method according to any one of claims 32 to 41, wherein the LY75 inhibitor comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within an LY75 genomic nucleic acid molecule.

45. The method according to claim 44, wherein the Cas protein is Cas9 or Cpf1.

46. The method according to claim 44 or claim 45, wherein the LY75 gRNA recognition sequence is located within SEQ ID NO:1.

47. The method according to claim 44 or claim 45, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.

48. The method according to any one of claims 44 to 47, wherein the gRNA comprises from about 17 to about 23 nucleotides.

49. The method according to any one of claims 44 to 47, wherein the LY75 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs:152-171.

50. The method according to any one of claims 32 to 41, wherein the CD164 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to a CD164 nucleic acid molecule.

51. The method according to claim 50, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).

52. The method according to any one of claims 32 to 41, wherein the CD164 inhibitor comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within a CD164 genomic nucleic acid molecule.

53. The method according to claim 52, wherein the Cas protein is Cas9 or Cpf1.

54. The method according to claim 52 or claim 53, wherein the CD164 gRNA recognition sequence is located within SEQ ID NO:55.

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55. The method according to claim 52 or claim 53, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.

56. The method according to any one of claims 52 to 55, wherein the gRNA comprises
5 from about 17 to about 23 nucleotides.

57. The method according to any one of claims 52 to 55, wherein the CD164 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs:172-191.

58. The method according to any one of claims 32 to 41, wherein the PARP1 inhibitor
10 comprises an inhibitory nucleic acid molecule that hybridizes to a PARP1 nucleic acid molecule.

59. The method according to claim 58, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).

60. The method according to any one of claims 32 to 41, wherein the PARP1 inhibitor
15 comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within a PARP1 genomic nucleic acid molecule.

61. The method according to claim 60, wherein the Cas protein is Cas9 or Cpf1.

62. The method according to claim 60 or claim 61, wherein the PARP1 gRNA recognition sequence is located within SEQ ID NO:113.

20 63. The method according to claim 60 or claim 61, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.

64. The method according to any one of claims 60 to 63, wherein the gRNA comprises from about 17 to about 23 nucleotides.

25 65. The method according to any one of claims 60 to 63, wherein the PARP1 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs:192-211.

66. A method of identifying a subject having an increased risk of developing clonal hematopoiesis of indeterminate potential (CHIP), the method comprising:

30 determining or having determined the presence or absence of an LY75 variant nucleic acid molecule, a CD164 variant nucleic acid molecule, and/or a PARP1 variant nucleic acid molecule in a biological sample obtained from the subject;

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wherein:

when the subject is LY75 reference, CD164 reference, and/or PARP1 reference, then the subject has an increased risk of developing CHIP; and

5 when the subject is heterozygous or homozygous for the LY75 variant nucleic acid molecule, the CD164 variant nucleic acid molecule, and/or the PARP1 variant nucleic acid molecule, then the subject has a decreased risk of developing CHIP.

67. The method according to claim 66, wherein the LY75 variant nucleic acid molecule, CD164 variant nucleic acid molecule, or PARP1 variant nucleic acid molecule is a missense variant, splice-site variant, a stop-gain variant, a start-loss variant, a stop-loss variant, a
10 frameshift variant, or an in-frame indel variant, or a variant that encodes a truncated predicted loss-of-function polypeptide.

68. The method according to claim 67, wherein the LY75 variant nucleic acid molecule encodes a truncated LY75 predicted loss-of-function polypeptide, the CD164 variant nucleic acid molecule encodes a truncated CD164 predicted loss-of-function polypeptide, or the PARP1
15 variant nucleic acid molecule encodes a truncated PARP1 predicted loss-of-function polypeptide.

69. The method according to any one of claims 66 to 68, wherein the subject is at risk of developing a hematologic cancer, a myeloid neoplasia, a lymphoid neoplasia, an atherosclerotic cardiovascular disease, a coronary heart disease, a myocardial infarction, or severe calcified
20 aortic valve stenosis.

70. The method according to any one of claims 66 to 69, wherein the subject is LY75 reference, and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount, and is administered an LY75 inhibitor.

25 71. The method according to any one of claims 66 to 69, wherein the subject is CD164 reference, and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount, and is administered a CD164 inhibitor.

72. The method according to any one of claims 66 to 69, wherein the subject is PARP1
30 reference, and the subject is administered or continued to be administered the therapeutic agent that prevents or reduces development of CHIP in a standard dosage amount, and is administered a PARP1 inhibitor.

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73. The method according to any one of claims 66 to 69, wherein the subject is heterozygous for the LY75 variant nucleic acid molecule, and the subject is administered or continued to be administered a therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is
5 administered an LY75 inhibitor.

74. The method according to any one of claims 66 to 69, wherein the subject is heterozygous for the CD164 variant nucleic acid molecule, and the subject is administered or continued to be administered a therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is
10 administered a CD164 inhibitor.

75. The method according to any one of claims 66 to 69, wherein the subject is heterozygous for the PARP1 variant nucleic acid molecule, and the subject is administered or continued to be administered a therapeutic agent that prevents or reduces development of CHIP in an amount that is the same as or less than a standard dosage amount, and is
15 administered a PARP1 inhibitor.

76. The method according to claim 70 or claim 73, wherein the LY75 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to an LY75 nucleic acid molecule.

77. The method according to claim 76, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin
20 RNA (shRNA).

78. The method according to claim 70 or claim 73, wherein the LY75 inhibitor comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within an LY75 genomic nucleic acid molecule.

79. The method according to claim 78, wherein the Cas protein is Cas9 or Cpf1.

80. The method according to claim 78 or claim 79, wherein the LY75 gRNA recognition sequence is located within SEQ ID NO:1.

81. The method according to claim 78 or claim 79, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.

82. The method according to any one of claims 78 to 81, wherein the gRNA comprises from about 17 to about 23 nucleotides.

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83. The method according to any one of claims 78 to 81, wherein the LY75 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs: 152-171.

84. The method according to claim 71 or claim 74, wherein the CD164 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to a CD164 nucleic acid molecule.

85. The method according to claim 84, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).

86. The method according to claim 71 or claim 74, wherein the CD164 inhibitor comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within a CD164 genomic nucleic acid molecule.

87. The method according to claim 86, wherein the Cas protein is Cas9 or Cpf1.

88. The method according to claim 86 or claim 87, wherein the CD164 gRNA recognition sequence is located within SEQ ID NO:55.

89. The method according to claim 86 or claim 87, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.

90. The method according to any one of claims 86 to 89, wherein the gRNA comprises from about 17 to about 23 nucleotides.

91. The method according to any one of claims 86 to 89, wherein the CD164 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs:172-191.

92. The method according to claim 72 or claim 75, wherein the PARP1 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to a PARP1 nucleic acid molecule.

93. The method according to claim 92, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).

94. The method according to claim 72 or claim 75, wherein the PARP1 inhibitor comprises a Cas protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within a PARP1 genomic nucleic acid molecule.

95. The method according to claim 94, wherein the Cas protein is Cas9 or Cpf1.

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96. The method according to claim 94 or claim 95, wherein the PARP1 gRNA recognition sequence is located within SEQ ID NO:113.

97. The method according to claim 94 or claim 95, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition
5 sequence.

98. The method according to any one of claims 94 to 97, wherein the gRNA comprises from about 17 to about 23 nucleotides.

99. The method according to any one of claims 94 to 97, wherein the PARP1 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID
10 NOs:192-211.

100. A therapeutic agent that prevents or reduces development of clonal hematopoiesis of indeterminate potential (CHIP) for use in the prevention or reduction of CHIP in a subject identified as having:

an LY75 variant genomic nucleic acid molecule, a CD164 variant genomic nucleic acid
15 molecule, and/or a PARP1 variant genomic nucleic acid molecule;

an LY75 variant mRNA molecule, a CD164 variant mRNA molecule, and/or a PARP1 variant mRNA molecule; or

an LY75 variant cDNA molecule, a CD164 variant cDNA molecule, and/or a PARP1 variant cDNA molecule.

20 101. A Lymphocyte Antigen 75 (LY75) inhibitor for use in the prevention or reduction of clonal hematopoiesis of indeterminate potential (CHIP) in a subject that:

a) is reference for an LY75 genomic nucleic acid molecule, an LY75 mRNA molecule, or an LY75 cDNA molecule; or

b) is heterozygous for:

25 i) an LY75 variant genomic nucleic acid molecule;

ii) an LY75 variant mRNA molecule; or

iii) an LY75 variant cDNA molecule.

102. A Cluster of Differentiation 164 (CD164) inhibitor for use in the prevention or reduction of clonal hematopoiesis of indeterminate potential (CHIP) in a subject that:

30 a) is reference for a CD164 genomic nucleic acid molecule, a CD164 mRNA molecule, or a CD164 cDNA molecule; or

b) is heterozygous for:

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- i) a CD164 variant genomic nucleic acid molecule;
- ii) a CD164 variant mRNA molecule; or
- iii) a CD164 variant cDNA molecule.

103. A Poly(ADP-Ribose) Polymerase 1 (PARP1) inhibitor for use in the prevention or
5 reduction of clonal hematopoiesis of indeterminate potential (CHIP) in a subject that:

a) is reference for a PARP1 genomic nucleic acid molecule, a PARP1 mRNA molecule, or
a PARP1 cDNA molecule; or

b) is heterozygous for:

- i) a PARP1 variant genomic nucleic acid molecule;
- 10 ii) a PARP1 variant mRNA molecule; or
- iii) a PARP1 variant cDNA molecule.

104. The method according to claim 101, wherein the LY75 inhibitor comprises an inhibitory
nucleic acid molecule that hybridizes to an LY75 nucleic acid molecule.

105. The method according to claim 104, wherein the inhibitory nucleic acid molecule
15 comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin
RNA (shRNA).

106. The method according to claim 101, wherein the LY75 inhibitor comprises a Cas
protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within an LY75
genomic nucleic acid molecule.

20 107. The method according to claim 106, wherein the Cas protein is Cas9 or Cpf1.

108. The method according to claim 106 or claim 107, wherein the LY75 gRNA recognition
sequence is located within SEQ ID NO:1.

109. The method according to claim 106 or claim 107, wherein a Protospacer Adjacent
Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition
25 sequence.

110. The method according to any one of claims 106 to 109, wherein the gRNA comprises
from about 17 to about 23 nucleotides.

111. The method according to any one of claims 106 to 109, wherein the LY75 gRNA
recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs
30 152-171.

112. The method according to claim 102, wherein the CD164 inhibitor comprises an
inhibitory nucleic acid molecule that hybridizes to a CD164 nucleic acid molecule.

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113. The method according to claim 112, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).

114. The method according to claim 102, wherein the CD164 inhibitor comprises a Cas
5 protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within a CD164 genomic nucleic acid molecule.

115. The method according to claim 114, wherein the Cas protein is Cas9 or Cpf1.

116. The method according to claim 114 or claim 115, wherein the CD164 gRNA recognition sequence is located within SEQ ID NO:55.

10 117. The method according to claim 114 or claim 115, wherein a Protospacer Adjacent Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.

118. The method according to any one of claims 114 to 117, wherein the gRNA comprises from about 17 to about 23 nucleotides.

15 119. The method according to any one of claims 114 to 117, wherein the CD164 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID NOs:172-191.

120. The method according to claim 103, wherein the PARP1 inhibitor comprises an inhibitory nucleic acid molecule that hybridizes to a PARP1 nucleic acid molecule.

20 121. The method according to claim 120, wherein the inhibitory nucleic acid molecule comprises an antisense nucleic acid molecule, a small interfering RNA (siRNA), or a short hairpin RNA (shRNA).

122. The method according to claim 103, wherein the PARP1 inhibitor comprises a Cas
25 protein and guide RNA (gRNA) that hybridizes to a gRNA recognition sequence within a PARP1 genomic nucleic acid molecule.

123. The method according to claim 122, wherein the Cas protein is Cas9 or Cpf1.

124. The method according to claim 122 or claim 123, wherein the PARP1 gRNA recognition sequence is located within SEQ ID NO:113.

125. The method according to claim 122 or claim 123, wherein a Protospacer Adjacent
30 Motif (PAM) sequence is about 2 to about 6 nucleotides downstream of the gRNA recognition sequence.

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126. The method according to any one of claims 122 to 125, wherein the gRNA comprises from about 17 to about 23 nucleotides.

127. The method according to any one of claims 122 to 125, wherein the PARP1 gRNA recognition sequence comprises a nucleotide sequence according to any one of SEQ ID

5 NOs:192-211.

128. A method of identifying a subject at risk of developing lung cancer, the method comprising:

determining or having determined the presence or absence of one or more clonal hematopoiesis of indeterminate potential (CHIP) somatic mutations in DNA Methyltransferase 3 Alpha (DNMT3A) and/or ASXL Transcriptional Regulator 1 (ASXL1) in a biological sample obtained from the subject;

wherein:

when the subject has a CHIP somatic mutation in DNMT3A and/or ASXL1, the subject has an increased risk of developing lung cancer; and

when the subject does not have a CHIP somatic mutation in DNMT3A and/or ASXL1, the subject does not have an increased risk of developing lung cancer.

129. The method according to claim 128, wherein the lung cancer comprises a non-small cell lung cancer, a small cell lung cancer, mesothelioma, lung carcinoid tumor, or a chest wall tumor.

130. The method according to claim 128 or claim 129, wherein the CHIP somatic mutation is in DNMT3A.

131. The method according to claim 128 or claim 129, wherein the CHIP somatic mutation is in ASXL1.

132. The method according to any one of claims 128 to 131, wherein the CHIP somatic mutation is a missense mutation, a splice-site mutation, a stop-gain mutation, a start-loss mutation, a stop-loss mutation, a frameshift mutation, or an in-frame indel mutation, or a mutation that results in a truncated predicted loss-of-function polypeptide.

133. The method according to any one of claims 128 to 132, wherein when the subject has one or more CHIP somatic mutations in DNMT3A and/or ASXL1, the subject is subjected to enhanced monitoring, a lifestyle change, and/or lowering exposure to a hazardous substance.

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134. The method according to claim 133, wherein the lifestyle change comprise cessation of smoking, and lowering exposure to an environmental risk factor selected from second-hand smoke, radon, and workplace smoke exposure.

135. The method according to claim 133, wherein the hazardous substance is selected from
5 asbestos, arsenic, nickel, chromium, beryllium, cadmium, silica, diesel exhaust, tar, and/or soot.

136. The method according to any one of claims 128 to 135, wherein when the subject has one or more CHIP somatic mutations in DNMT3A and/or ASXL1, the subject is administered a therapeutically effective amount of elrotinib, 5-(p-methoxyphenyl)-1,2-dithiole-3-thione, deguelin, or iloprost, or any combination thereof.

10 137. The method according to any one of claims 128 to 135, further comprising determining whether the subject carries a DNMT3A R882H somatic mutation, wherein when the subject carries the DNMT3A R882H somatic mutation, the subject is excluded from treatment with a PARP1 inhibitor.

138. The method according to any one of claims 128 to 135, further comprising determining
15 whether the subject carries a TET2 somatic mutation deficiency, wherein when the subject carries the TET2 somatic mutation deficiency, the subject is excluded from treatment with a PARP1 inhibitor.

139. A method of stratifying a suitable lung cancer patient for treatment with a PARP1 inhibitor, the method comprising determining whether the patient carries a DNMT3A R882H
20 somatic mutation or TET2 somatic mutation deficiency, wherein the patient with the DNMT3A R882H somatic mutation or TET2 somatic mutation deficiency is excluded from a PARP1 inhibitor treatment regimen.

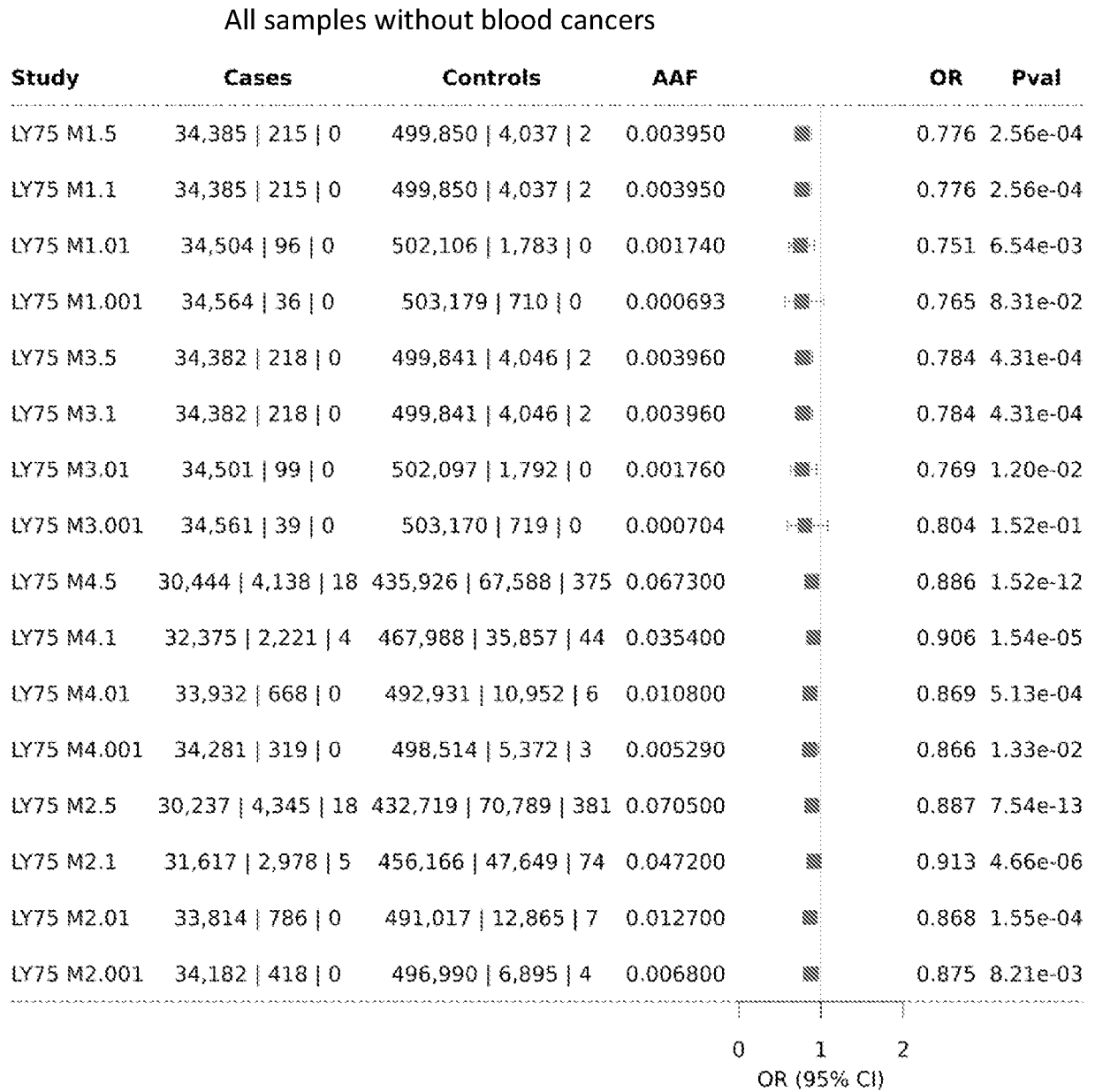


Figure 1

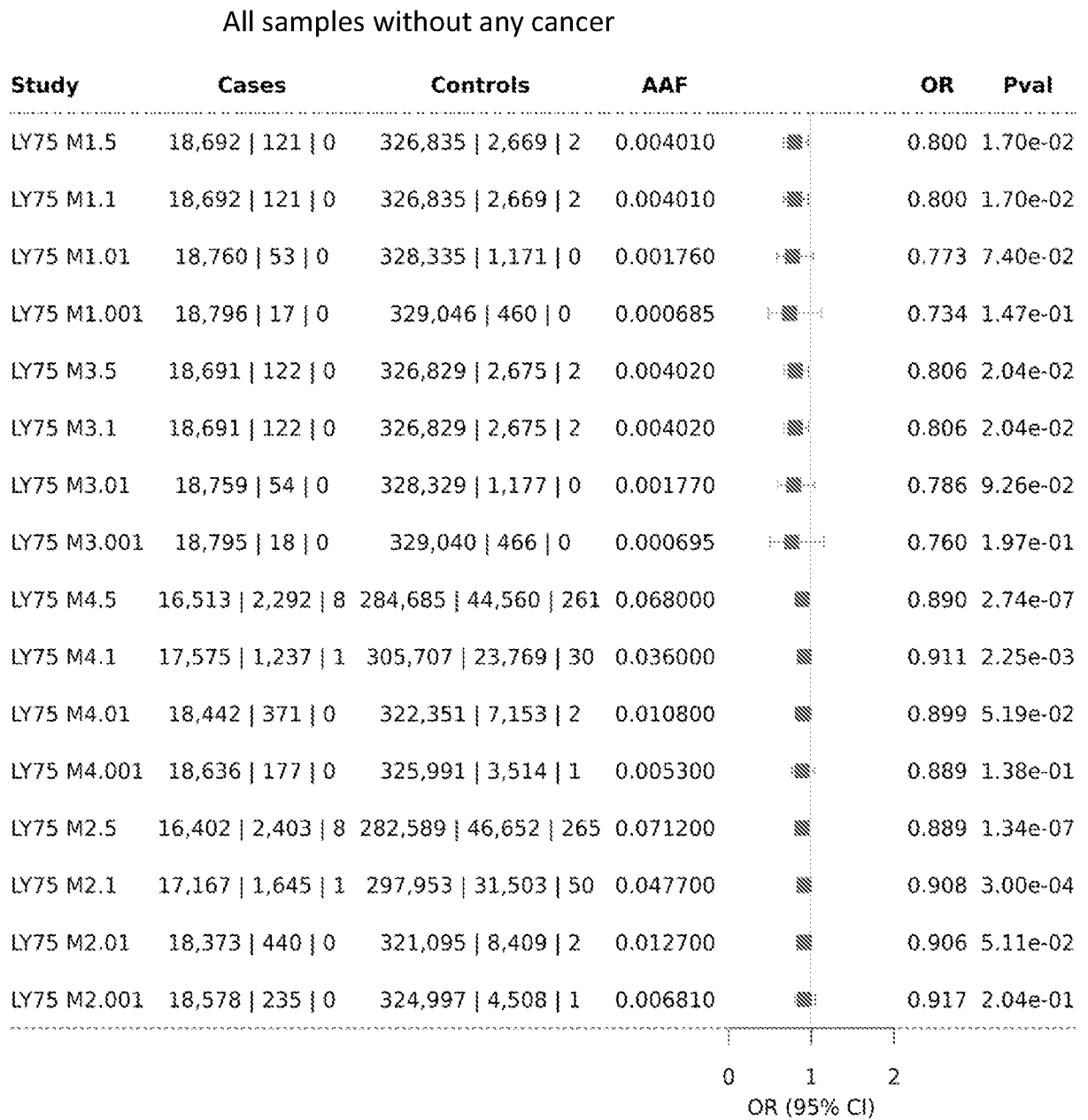


Figure 1 (cont.)

2 159834145 G A

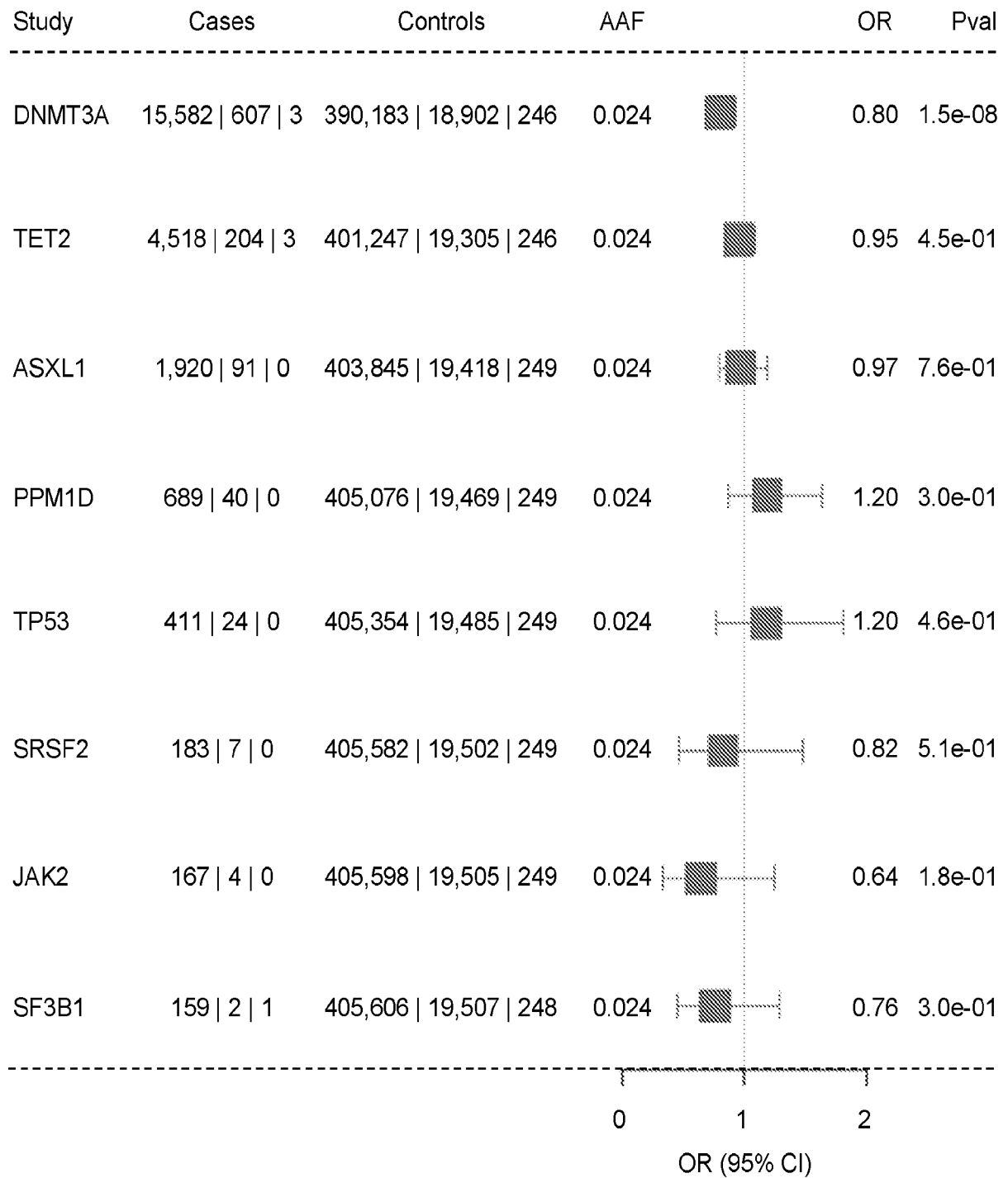


Figure 2

M2.5

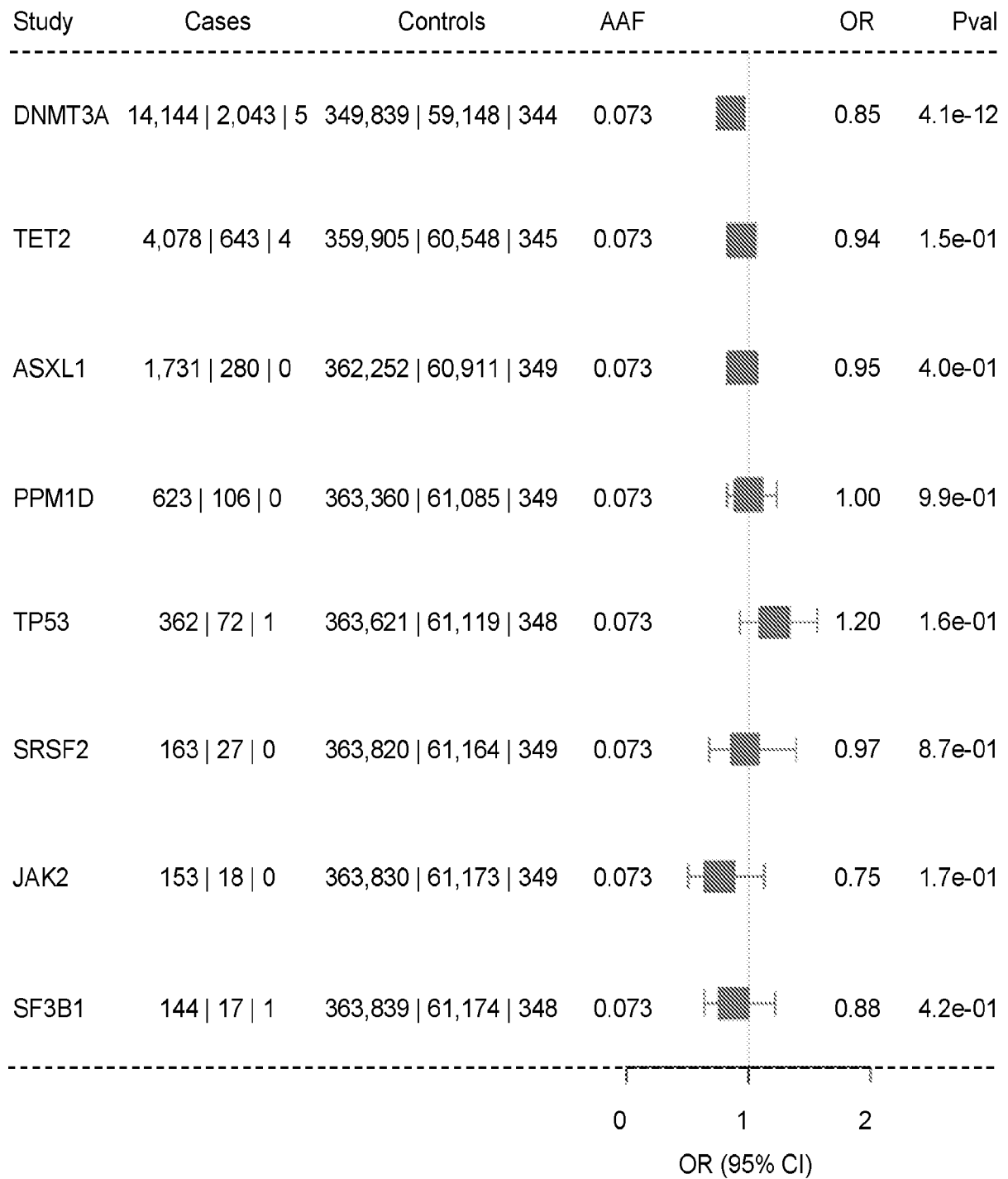


Figure 2 (cont.)

M1.1

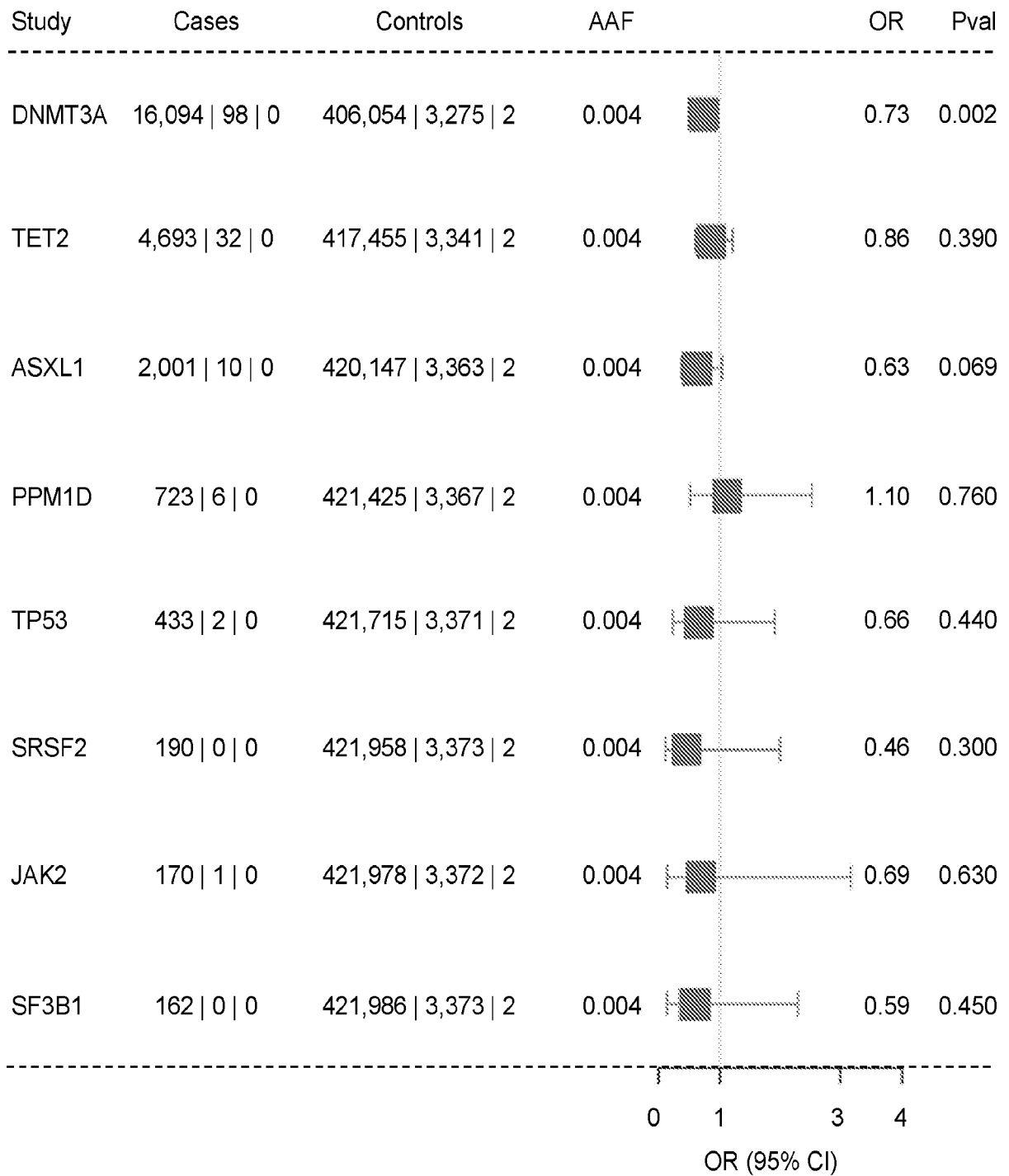


Figure 2 (cont.)

Chip Minus Malignant Blood LY75-CD302 Masks

Study	Cases	Controls	AAF	OR	Pval
LY75-CD302 M1.5	34,362 238 0	499,548 4,339 2	0.004250	0.801	7.99e-04
LY75-CD302 M1.1	34,362 238 0	499,548 4,339 2	0.004250	0.801	7.99e-04
LY75-CD302 M1.01	34,481 119 0	501,802 2,087 0	0.002050	0.805	2.28e-02
LY75-CD302 M1.001	34,551 49 0	502,928 961 0	0.000938	0.772	5.16e-02
LY75-CD302 M3.5	34,346 254 0	499,385 4,502 2	0.004420	0.817	1.76e-03
LY75-CD302 M3.1	34,346 254 0	499,385 4,502 2	0.004420	0.817	1.76e-03
LY75-CD302 M3.01	34,465 135 0	501,639 2,250 0	0.002210	0.842	4.88e-02
LY75-CD302 M3.001	34,539 61 0	502,800 1,089 0	0.001070	0.819	1.06e-01
LY75-CD302 M4.5	30,526 4,056 18	437,443 66,073 373	0.065800	0.888	6.48e-12
LY75-CD302 M4.1	32,464 2,132 4	469,578 34,269 42	0.033900	0.909	3.97e-05
LY75-CD302 M4.01	33,876 724 0	492,013 11,870 6	0.011700	0.873	5.09e-04
LY75-CD302 M4.001	34,235 365 0	497,617 6,269 3	0.006170	0.851	2.89e-03
LY75-CD302 M2.5	30,310 4,272 18	434,074 69,436 379	0.069200	0.889	2.59e-12
LY75-CD302 M2.1	31,697 2,898 5	457,574 46,243 72	0.045800	0.914	9.09e-06
LY75-CD302 M2.01	33,749 851 0	489,948 13,934 7	0.013700	0.872	1.36e-04
LY75-CD302 M2.001	34,135 465 0	496,094 7,791 4	0.007670	0.864	2.37e-03

012

OR (95% CI)

Figure 3

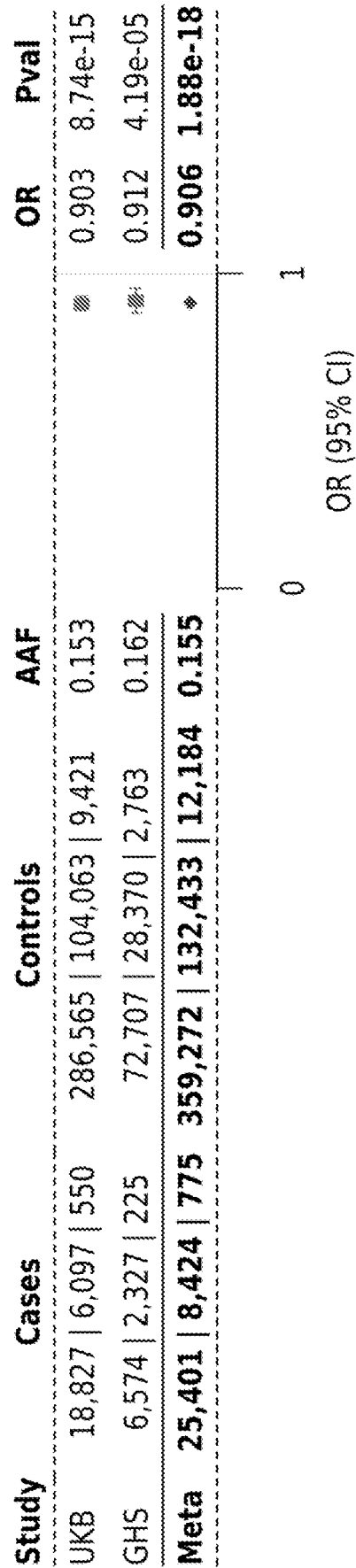


Figure 4

uid	geneName	Pval	Dataset	Cis_Trans	Effect_allele	Ref_allele	Effect
1:226367601:A:G	PARP1	7.2e-17	Fehrmann_Whole_Blood	CIS	G	A	-8.34
1:226388569:A:G	PARP1	1.23591e-15	Brown_LCLs	CIS	NA	NA	NA
1:226402257:G:A	PARP1	3.48e-12	Nedelec_Macrophages_salmonella_asQTL	CIS	NA	NA	NA
1:226407676:C:G	PARP1	1.09e-13	Nedelec_Macrophages_listeria_asQTL	CIS	NA	NA	NA
1:226407676:C:G	PARP1	1.4199999999999998e-31	Nedelec_Macrophages_baseline_asQTL	CIS	NA	NA	NA
1:226408985:T:A	PARP1	2.9335999999999998e-195	Vosa_Whole_Blood	CIS	A	T	-29.8094
1:226414809:T:C	PARP1	8.88e-16	Hao_Lung	CIS	C	T	-8.06
1:226421638:A:C	PARP1	5.79546e-40	Zhernakova_Whole_Blood_gene	CIS	C	A	-13.2311
1:226422811:C:G	PARP1	2.22638e-17	Joehanes_Whole_Blood	CIS	C	G	0.0252

Figure 5

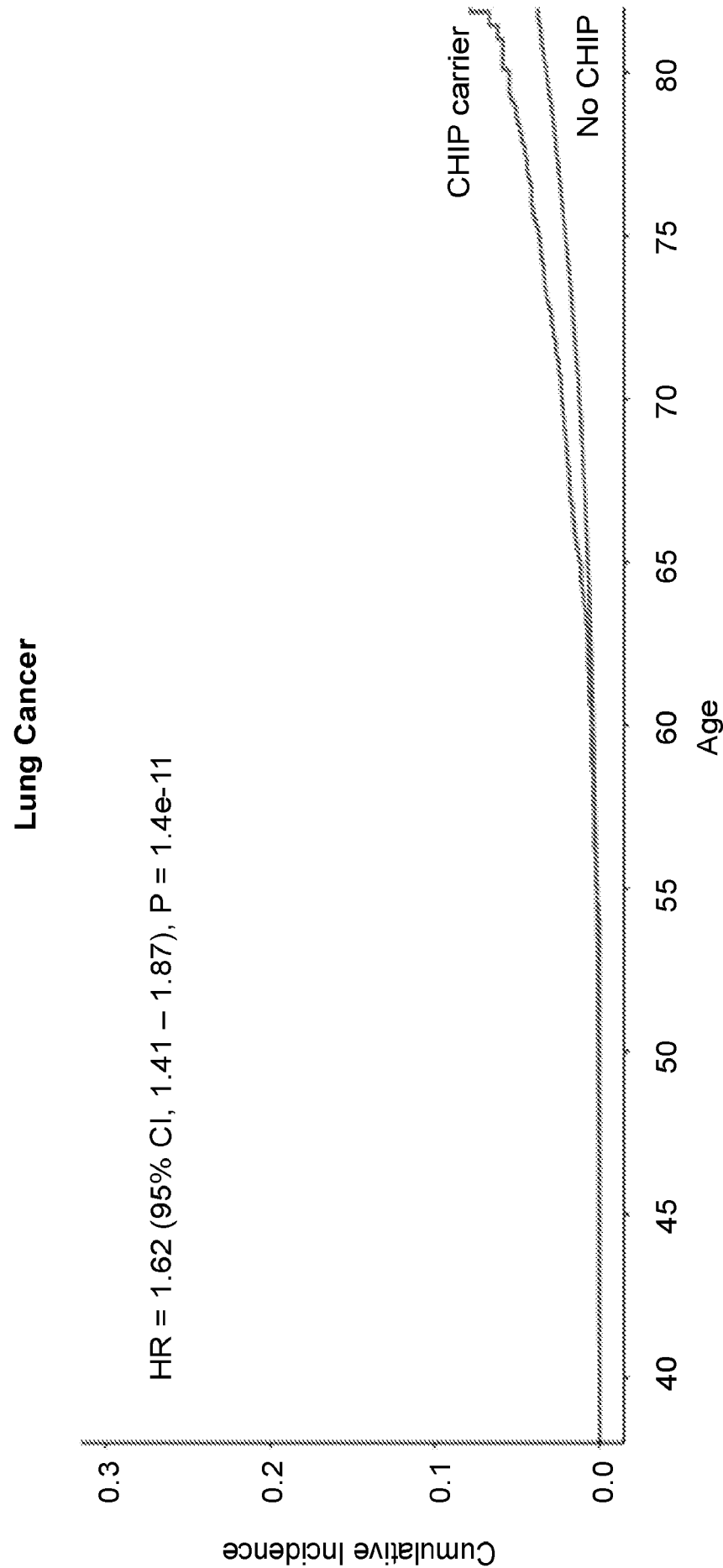


Figure 6

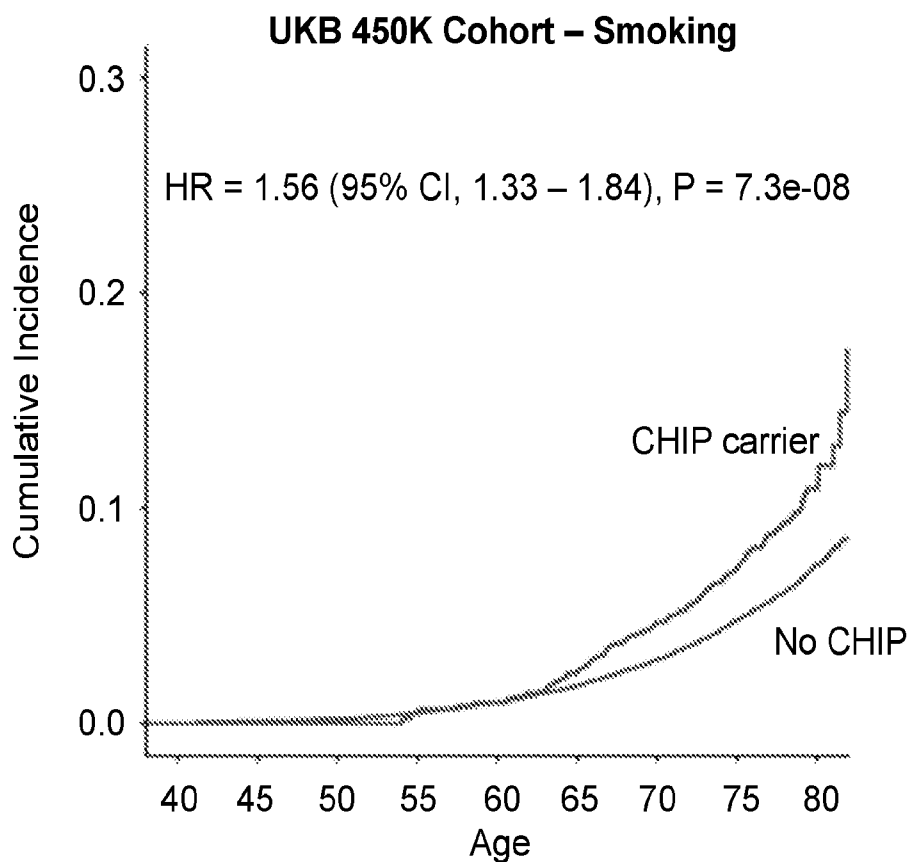
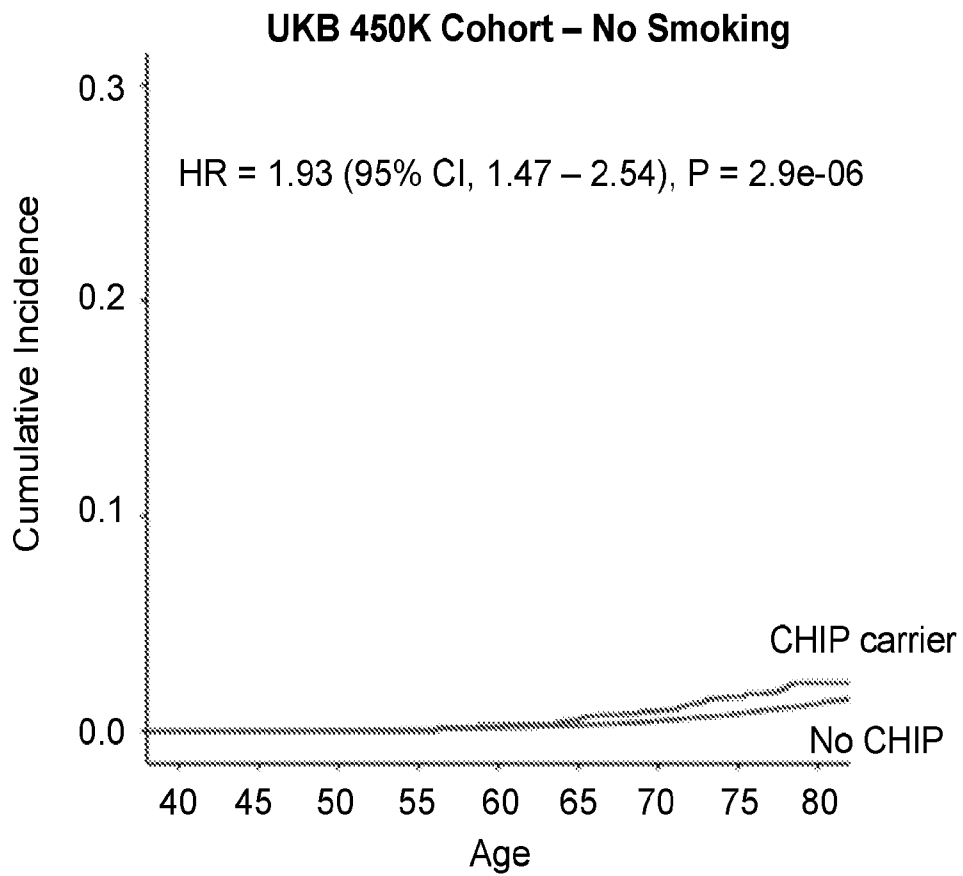


Figure 6 (cont.)

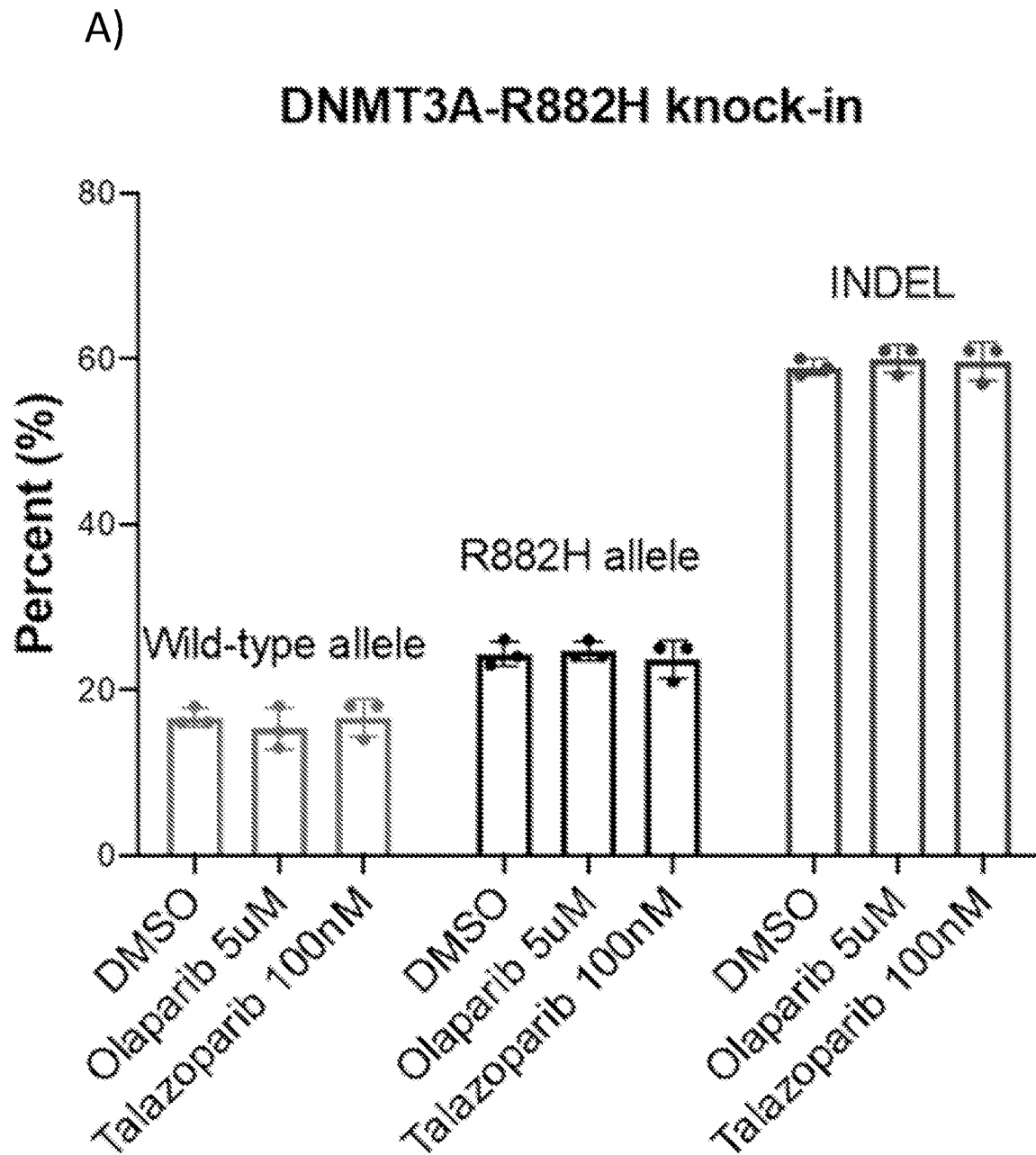


Figure 7

B)

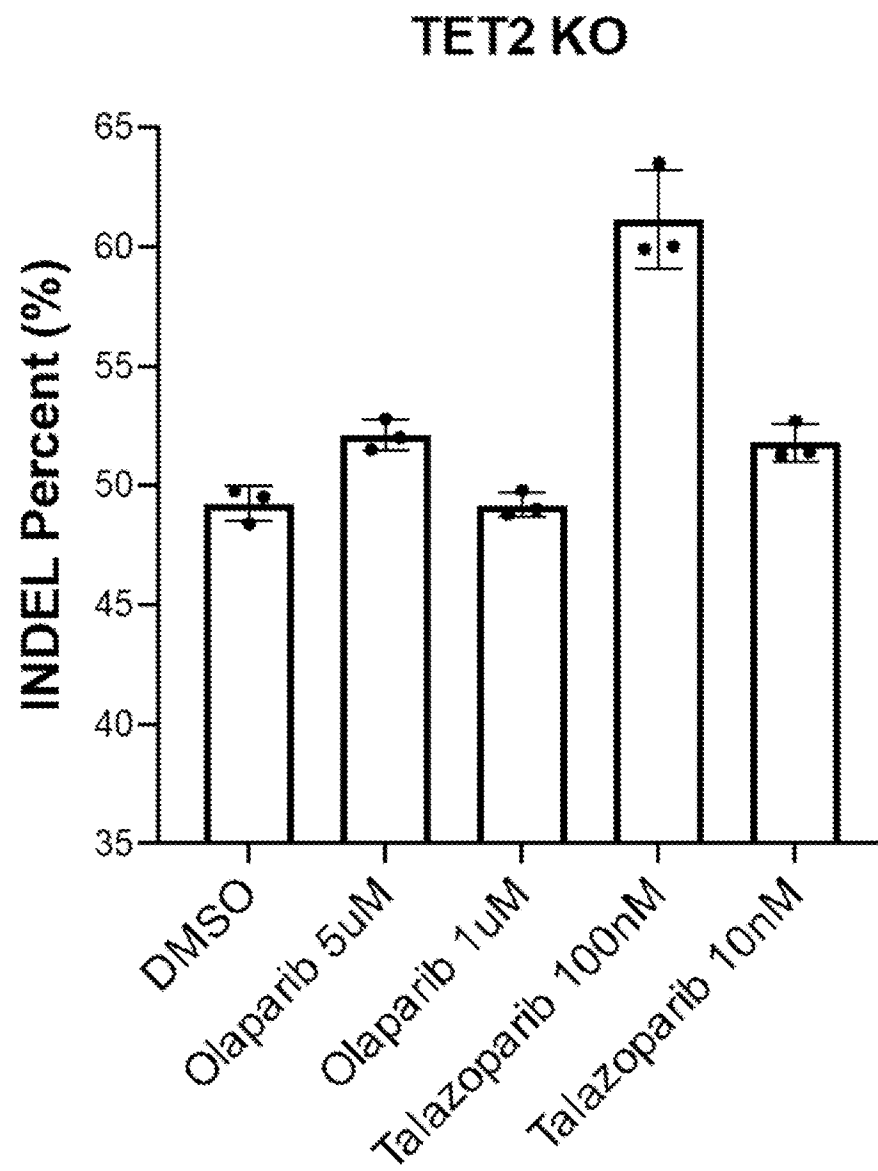
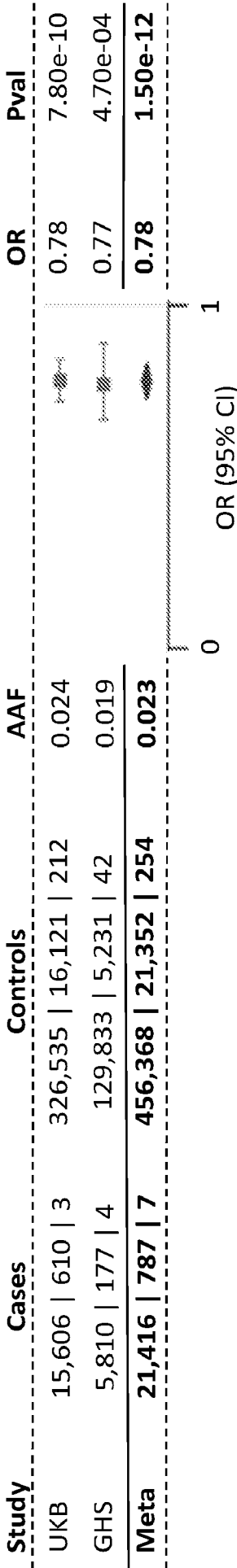


Figure 7 (cont.)

LY75 rs78446341-A



LY75 rs147820690-T

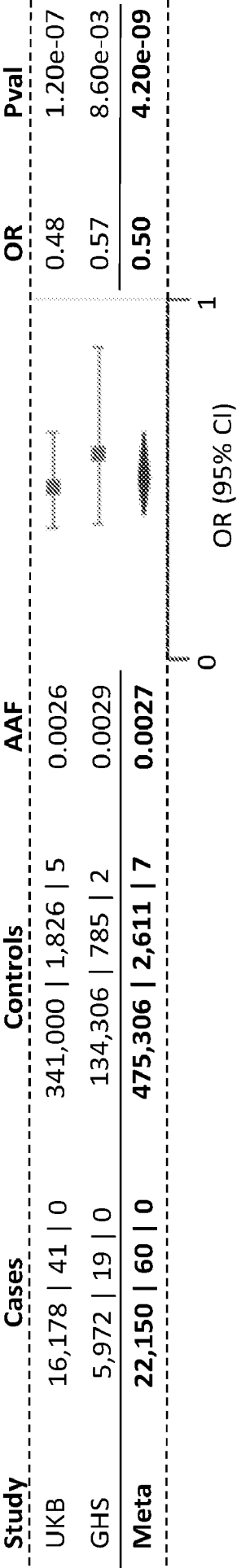


Figure 8