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(54) Title: METHODS OF PREDICTING THE DEVELOPMENT OF COMPLEMENT-MEDIATED DISEASE

(57) Abstract: Disclosed herein are methods and compositions for the diagnosis and treatment of complement-mediated disease.



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METHODS OF PREDICTING THE DEVELOPMENT OF COMPLEMENT-MEDIATED DISEASE

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5 EY017404 awarded by the NIH. The government has certain rights in the invention.

CROSS-REFERENCES TO RELATED APPLICATIONS

[0002] This application claims benefit of U.S. Provisional Patent Application No.
61/480,929, filed April 29, 2011. The entire contents of U.S. Provisional Patent
Application No. 61/480,929 are incorporated herein by reference.

FIELD OF THE INVENTION

[0003] The invention relates generally to the diagnosis and treatment of complement-
mediated disease in a human subject. Specifically, the invention relates to the
prediction or diagnosis of age-related macular degeneration through the detection of a
collection of polymorphisms and haplotypes comprised of multiple variations in the
15 CFH-to-F13B locus.

BACKGROUND

[0004] Age related macular degeneration (AMD), a complement-mediated disease, is
the leading cause of blindness in the elderly, a problem that will grow worse with an
aging population. Accurate identification of risk for developing complement-
20 mediated diseases such as AMD is essential for both starting treatment early in
disease progression and identifying causal genetic variants to better target therapeutic
strategies and development. Previously, loci have been implicated in risk for AMD
on Chr1, Chr10, Chr6 (CFB), and Chr19 (C3).

[0005] The RCA gene cluster, sometimes referred to below as the “CFH-to-F13B
25 locus,” is located on chromosome 1q32 and includes the genes for complement factor
H (CFH), five Factor H-related genes (CFHR1, CFHR2, CFHR3, CFHR4 and
CFHR5), and the gene encoding the beta subunit of coagulation factor XIII. See US
Pat. Pub. US 2007/0020647, incorporated herein by reference). CFH is a significant

regulator of complement activity and is thought to be essential to prevent injury to self-tissues by inappropriate C3 activation. Additionally, alterations in CFH activity have been associated with altered binding to membrane bound glycoproteins, other complement inhibitors, and the surface of pathogenic bacteria.

5 [0006] Previously, haplotype studies have focused on CFH and only rarely extended as far as CFHR2. However, the full CFH locus is an area of the genome that contains additional variants that associate with complement-mediated diseases such as AMD. Therefore, what is needed are more effective methods of describing risk for having or developing complement-mediated disease at this locus. Additionally, what is needed
10 are methods to place known risk and protective variants into the broader genetic background in which they exist, that is the entire CHF-to-F13B locus.

SUMMARY

[0007] Described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising
15 determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates the subject's susceptibility for
20 having or developing a complement-mediated disease.

[0008] Also described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A,
25 H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates the subject's susceptibility for having or developing a complement-mediated disease.

[0009] Also described herein are methods for determining a Caucasian subject's
30 susceptibility to having or developing a complement-mediated disease comprising

determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates the subject's susceptibility for having or developing a complement-mediated disease.

[0010] Also described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus, wherein the SNPs are: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of at least six of the SNPs indicates the subject's susceptibility for having or developing a complement-mediated disease.

[0011] Also described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus, wherein the SNPs is: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of one or more of the SNPs indicates the subject's susceptibility for having or developing a complement-mediated disease.

[0012] Also described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus, wherein the SNP is: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939, and wherein the presence of the SNP indicates the subject's susceptibility for having or developing a complement-mediated disease.

[0013] Also described herein are methods of identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A,

H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of treatment for a complement-mediated disease.

5 [0014] Also described herein are methods of identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, 10 H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of treatment for a complement-mediated disease.

[0015] Also described herein are methods of identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the 15 Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is 20 in need of treatment for a complement-mediated disease.

[0016] Also described herein are methods of identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus, wherein the SNPs are: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, 25 rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of at least six of the SNPs indicates whether the subject is in need of treatment for a complement-mediated disease.

[0017] Also described herein are methods of identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the 30 subject the identity of one or more SNPs in the CFH-to-F13B locus, wherein the SNPs is: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or

(ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of one or more of the SNPs indicates whether the subject is in need of treatment for a complement-mediated disease.

[0018] Also described herein are methods of identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus, wherein the SNPs are: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939, and wherein the presence of one or more of the SNPs indicates whether the subject is in need of treatment for a complement-mediated disease.

[0019] Also described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of prophylactic treatment for a complement-mediated disease.

[0020] Also described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of prophylactic treatment for a complement-mediated disease.

[0021] Also described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B,

H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of prophylactic treatment for a complement-mediated disease.

[0022] Also described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of prophylactic treatment for a complement-mediated disease.

[0023] Also described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for a complement-mediated disease comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus, wherein the SNPs is: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of one or more of the SNPs indicates whether the subject is in need of prophylactic treatment for a complement-mediated disease.

[0024] Also described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for a complement-mediated disease comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus, wherein the SNPs are: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939, and wherein the presence of one or more of the SNPs indicates whether the subject is in need of prophylactic treatment for a complement-mediated disease.

[0025] Also described herein are methods of identifying a Caucasian subject appropriate for an a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A,

H13_62_A, H14_62_A, H15_62_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is appropriate for the clinical trial.

[0026] Also described herein are methods of identifying a Caucasian subject appropriate for an a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is appropriate for the clinical trial.

[0027] Also described herein are methods of identifying a Caucasian subject appropriate for an a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is appropriate for the clinical trial.

[0028] Also described herein are methods of identifying a Caucasian subject appropriate for an a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus, wherein the SNPs are: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of at least six of the SNPs indicates whether the subject is appropriate for the clinical trial.

[0029] Also described herein are methods of identifying a Caucasian subject appropriate for an a complement-mediated disease clinical trial comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus, wherein the SNPs is: (i) rs1061170, rs1410996, rs2274700, rs3753395,

rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of one or more of the SNPs indicates whether the subject is appropriate for the clinical trial.

[0030] Also described herein are methods of identifying a Caucasian subject appropriate for an a complement-mediated disease clinical trial comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus, wherein the SNPs are: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939, and wherein the presence of one or more of the SNPs indicates whether the subject is appropriate for the clinical trial.

[0031] Also described herein are kits kit comprising an assay for detecting one or more haplotypes in a nucleic acid sample of a subject, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof.

[0032] Also described herein are methods for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates the subject's susceptibility for having or developing age-related macular degeneration.

[0033] Also described herein are methods of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates whether the subject is in need of treatment for age-related macular degeneration.

[0034] Also described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

[0035] Also described herein are methods of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates whether the subject is appropriate for the clinical trial.

BRIEF DESCRIPTION OF THE FIGURES

[0036] The accompanying figures, which are incorporated in and constitute a part of this specification, illustrate several aspects and together with the description serve to explain the principles of the invention.

5 [0037] Figure 1 shows major haplotypes H1_61_A through H15_62_A.

[0038] Figure 2 shows the 63 SNPs utilized in the preliminary haplotype studies.

[0039] Figure 3 shows minor haplotypes H16_62_A through H163_62_A.

[0040] Figure 4 shows the verification of haplotypes derived from the GSH – 1073 cohort and the GSH – 927 cohort.

10 [0041] Figure 5 shows the single SNP association data for the 62 SNP panel in the GSH cohort.

[0042] Figure 6 shows disease associations that were analyzed for the entire extended haplotypes using Haploview 4.2.

[0043] Figure 7 shows the haplotype tagging SNPs for 62 SNP haplotypes.

15 [0044] Figure 8 shows the 51_A haplotype tagging SNPs (51_A htSNPs).

[0045] Figure 9 shows the 51_B haplotype tagging SNPs (51_B htSNPs).

[0046] Figure 10 shows the effect of generating haplotypes using fewer and fewer SNPs has on association values for haplotypes.

20 [0047] Figure 11 shows the qPCR assay for R3-1 deletion, regular PCR assay for homozygous deletion or non-deletion.

[0048] Figure 12 shows the assessment of 51 SNP haplotypes in the Sib-pair cohort.

[0049] Figure 13 shows the 62 single marker (SNPs) and haplotypes association with gender in the GSH case-control cohort the male and female gender association in the '927 cohort.

[0050] Figure 14 shows the haplotype boundaries extended beyond that covered by 63 SNPs. Provided in the figure is a list of all Hapmap SNPs.

[0051] Figure 15 shows the CFH locus and the haplotypes in the 51_B cohort.

[0052] Figure 16 shows a comparison of 51_A and 62_A haplotypes.

5 [0053] Figure 17 shows shows the frequencies of the 62_A haplotypes.

[0054] Figure 18 shows the 62_A SNP haplotype schema regrouped with P values.

[0055] Figure 19 shows a comparison of the 51 SNP haplotypes in the 1073 cohort and combined cohorts.

[0056] Figure 20 shows the '927 cohort and the 62_A to 51_A conversion.

10 [0057] Figure 21 shows a haplotype tree for the 62_A cohort. Figure 21(A) shows the hierarchical relationships between the 163 62_A SNP-based haplotypes.

[0058] Figure 22 shows the association of haplotypes in the '927 cohort using a 51 SNP-based haplotype.

[0059] Figure 23 shows the gender associations of the 62_A haplotypes.

15 [0060] Figure 24 shows the gender associations of the 51_B haplotypes.

[0061] Figure 25 shows haplotype disease association.

[0062] Figure 26 shows haplotype disease association based on gender.

[0063] Figure 27 shows the SNPs used in a haplotype reconstruction.

[0064] Figure 28 shows Taqman qPCR copy number variation data.

20 [0065] Figure 29 shows the frequency of subjects with varying number of SNPs screened.

[0066] Figure 30 shows a deletion decision tree made to predict deletion status from three SNPs.

[0067] Figure 31 shows the results of the real-time quantitative PCR (qPCR) used to determine the copy number state of the CFHR3 and CFHR1 genes.

[0068] Figure 32 shows the H1-H19 haplotypes based on 1073 cohort screened for 63 SNPs.

5 [0069] Figure 33 shows haplotype frequencies calculated on 136 unrelated UGRP individuals, who were mostly first generation grandparents, using Haploview program's phased haplotype input mode, Haps format.

[0070] Figure 34 shows association of 63 SNP-based CFH-to-F13B Haplotypes with AMD.

10 [0071] Figure 35 shows the diplotype analysis of the overall 51_B cohort.

[0072] Figure 36 shows diplotype analysis for males in the 51_B cohort.

[0073] Figure 37 shows diplotype analysis for females in the 51_B cohort.

[0074] Figure 38 shows the combined cohort with all haplotypes, 0vs1b-4, highlighted to match major HapMap haplotypes.

15 [0075] Figure 39 shows the major CFH-to-F13B HapMap haplotypes with >0.5% frequencies for CEU.

[0076] Figure 40 shows the marker-rs number IDs shown in Figure 39.

[0077] Figure 41 shows a comparison of the 51_B-based haplotypes to HapMap haplotypes >0.05% frequencies.

20 [0078] Figure 42 shows single marker associations; Controls (0) vs all AMD in the 51_B cohort.

[0079] Figure 43 shows single marker associations; Controls (0) vs early AMD (1b-3) in the 51_B cohort.

25 [0080] Figure 44 shows single marker associations; Controls (0) vs geographic atrophy (4A) in the 51B cohort.

[0081] Figure 45 shows the single marker associations; Controls (0) vs CNV (4B) in the 51_B cohort.

[0082] Figure 46 shows the haplotype associations; Controls (0) vs all AMD, early AMD (1B-3), GA only (4A) & CNV only (4B) in the 51_B cohort.

5 [0083] Figure 47 shows the diplotype associations; Controls (0) vs early AMD (1B-3) in the 51_B cohort.

[0084] Figure 48 shows the diplotype associations; Controls (0) vs CNV (4B) in the 51_B cohort.

10 [0085] Additional advantages of the invention will be set forth in part in the description which follows, and in part will be obvious from the description, or can be learned by practice of the invention. The advantages of the invention will be realized and attained by means of the elements and combinations particularly pointed out in the appended claims. It is to be understood that both the foregoing general
15 and are not restrictive of the invention, as claimed.

DESCRIPTION

[0086] The present invention can be understood more readily by reference to the following detailed description of the invention and the Examples included therein.

20 [0087] 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 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-express basis for interpretation, including matters of logic with respect to
25 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.

[0088] Definitions

[0089] As used in the specification and the appended claims, the singular forms “a,” “an” and “the” include plural referents unless the context clearly dictates otherwise. Thus, for example, reference to “a pharmaceutical carrier” includes mixtures of two or more such carriers, and the like.

5 [0090] Ranges can be expressed herein as from “about” one particular value, and/or to “about” another particular value. When such a range is expressed, another embodiment includes from the one particular value and/or to the other particular value. Similarly, when values are expressed as approximations, by use of the antecedent “about,” it will be understood that the particular value forms another
10 embodiment. It will be further understood that the endpoints of each of the ranges are significant both in relation to the other endpoint, and independently of the other endpoint. It is also understood that there are a number of values described herein, and that each value is also herein disclosed as “about” that particular value in addition to the value itself. For example, if the value “10” is disclosed, then “about 10” is also
15 disclosed. It is also understood that when a value is disclosed that “less than or equal to” the value, “greater than or equal to the value” and possible ranges between values are also disclosed, as appropriately understood by the skilled artisan. For example, if the value “10” is disclosed the “less than or equal to 10” as well as “greater than or equal to 10” is also disclosed. It is also understood that throughout the application,
20 data are provided in a number of different formats, and that these data, represent endpoints, starting points, and ranges for any combination of the data points. For example, if a particular data point “10” and a particular data point 15 are disclosed, it is understood that greater than, greater than or equal to, less than, less than or equal to, and equal to 10 and 15 are considered disclosed as well as between 10 and 15. It
25 is also understood that each unit between two particular units is also disclosed. For example, if 10 and 15 are disclosed, then 11, 12, 13, and 14 are also disclosed.

[0091] “Optional” or “optionally” means that the subsequently described event or circumstance may or may not occur and that the description includes instances where said event or circumstance occurs and instances where it does not.

30 [0092] The word “or” as used herein means any one member of a particular list and also includes any combination of members of that list.

[0093] “Complement-Mediated Disease” as used herein refers to a disease caused by or resulting from abnormal complement activation or function of its four major pathways. The complement pathway consists of three stages: recognition, enzymatic activation and membrane attack leading to cell death. “Complement-Mediated Disease” as used herein also refers to a disease caused by or resulting from alterations of the classical, exogenous, mannan-binding lectin (MBL) or alternative pathways. Examples of Complement-Mediated Disease include, but are not limited to, atypical haemolytic uraemic syndrome (aHUS), membranoproliferative glomerulonephritis type II (MPGN II; also known as dense deposit disease), systemic lupus erythematosus (SLE), pyogenic infections, hemolysis and thrombosis, partial lipodystrophy, hereditary angioedema, paroxysmal nocturnal haemoglobinuria (PNH), age-related macular degeneration (AMD), rheumatoid arthritis, myocarditis, multiple sclerosis, IgA nephropathy, Henoch–Schoenlein purpura, glaucoma, post-streptococcal disease, anti-phospholipid antibody syndrome, recurrent pregnancy loss, asthma, antibody-mediated cutaneous disease and anti-nuclear cytoplasmic antigen-associated pauci-immune vasculitis.

[0094] “Haplotype” as used herein refers to a DNA sequence or a combination of DNA sequences present at various loci on a chromosome that are transmitted together; a haplotype may be one locus, several loci, or an entire chromosome depending on the number of recombination events that have occurred between a given set of loci.

[0095] “Risk Haplotype” as used herein refers to a haplotype of a subject wherein the presence of the haplotype indicates the subject’s increased risk of developing a complement-mediated disease (e.g. AMD) when compared to a subject without said risk haplotype. A “Risk Haplotype” also refers to a haplotype that occurs more often in subjects with Complement-Mediated Disease (e.g. AMD) (cases) than in subjects without Complement-Mediated Disease (e.g. AMD) (controls) in a given cohort. In other words, the case versus control percentage is different in cases than in controls as illustrated in the tables herein (see FIG. 15). Although not required, the difference between the number of subjects with Complement-Mediated Disease (e.g. AMD) (case) and the number of subjects without Complement-Mediated Disease (e.g. AMD)

(control) can be statistically significant. For example, H2_62_A, H2_51_A, and H2_51_B are examples of “Risk Haplotypes” as they relate to AMD.

[0096] “Protective Haplotype” as used herein refers to a haplotype of a subject wherein the presence of the haplotype indicates the subject’s decreased risk of developing Complement-Mediated Disease (e.g. AMD) when compared to a subject without said protective haplotype. A “Protective Haplotype” also refers to a haplotype that occurs less often in subjects with Complement-Mediated Disease (e.g. AMD) (cases) than in subjects without Complement-Mediated Disease (e.g. AMD) (controls), in other words, the case versus control percentage is lower in cases than in controls as illustrated in the tables herein (See FIG. 15). Although not required, the difference between the number of subjects with Complement-Mediated Disease (e.g. AMD) (case) and the number of subjects without Complement-Mediated Disease (e.g. AMD) (control) can be statistically significant. For example, H5_62_A, H5_51_A, and H5_51_B are examples of “protective haplotypes” as they relate to AMD.

[0097] “Neutral Haplotype” as used herein refers to a haplotype of a subject wherein the presence of the haplotype does not indicate the subject’s increased or decreased risk of developing Complement-Mediated Disease (e.g. AMD). A “Neutral Haplotype” also refers to a haplotype that occurs about equally in subjects with Complement-Mediated Disease (e.g. AMD) (cases) and in subjects without Complement-Mediated Disease (e.g. AMD) (controls), in other words, the case versus control percentage is about equal in cases and controls as illustrated in the tables herein (See FIG. 15). Although not required, the difference between the number of subjects with Complement-Mediated Disease (e.g. AMD) (case) and the number of subjects without Complement-Mediated Disease (e.g. AMD) (control) can be non-statistically significant. For example, H4_62_A, H4_51_A, and H4_51_B are examples of “neutral haplotypes” as they relate to AMD.

[0098] Haplotypes as described herein can be referred to by the generic nomenclature: “Hx_Y_Z”, wherein “Hx” indicates the numbering of the specific haplotype as determined by the frequency of the specific haplotype in a given population or cohort. For example, H2 indicates a specific haplotype that occurs with the second highest frequency in the given data set, population or cohort. “Y”, in the

context of this nomenclature, specifies the SNP panel from which the overall haplotype was determined. For example, Hx_62_Z would refer to the 62 SNP Panel as described herein, whereas Hx_51_Z refers to the 51 SNP Panel as described herein. “Z”, in the context of this nomenclature, refers to the specific cohort used in
5 ascertaining the specific haplotype. For example, Hx_Y_A would refer to the ‘927 cohort’ as described herein, whereas Hx_Y_B refers to the combined cohort as described herein.

[0099] “Haplotype Tagging Haplotype” refers to haplotypes that can tag one or more of the haplotypes described herein. Such haplotypes can be referred to as “tagging
10 haplotypes”. Such tagging haplotypes can be identified based on HapMap Analysis as described herein. When such tagging haplotypes are referenced herein, the generic nomenclature: “Hx_Y_Z_t” can be used wherein “t” represents a “tagging haplotype” and Hx, Y, and Z represent the same categories as described above for the generic nomenclature “Hx_Y_Z”. For example, H2_62_A_1 would represent a first
15 haplotype that tags the H2_62_A haplotype.

[00100] “Major Haplotype” as used herein refers to a haplotype that occurs in greater than 1% of the population, data set or cohort being examined.

[00101] “Minor Haplotype” as used herein refers to a haplotype that occurs in 1% or less than 1% of the population, data set or cohort being examined.

[00102] “CFH-to-F13B Locus” as used herein refers to a region of chromosome
20 1q25-q31 (approximately 196,580,000 to 197,053,000; hg19) containing the following genes and pseudogenes: complement factor H (CFH) and its truncated isoform (CFHT), the complement factor H related genes 3, 1, 4, 2 and 5 (CFHR3, CFHR1, CFHR4, CFHR2 and CFHR5), a CFHR pseudogene () and factor 13B
25 (F13B). The CFH-to-F13B Locus includes all non-genic DNA, including that extending proximal to CFH and distal to F13B. Blocks of linkage disequilibrium (LD) that initiate within the CFH-to-F13B and extend proximal to CFH and/or distal to F13b are also included.

[00103] “62 SNP Panel” as used herein refers to 62 SNPs that were selected based on
30 their significance of association as well as locations selected to provide coverage

across the entire CFH-to-F13B Locus and its LD blocks. The 62 SNP Panel consists of the SNPs shown in FIG. 17.

[00104] “51 SNP Panel” as used herein refers to 51 SNPs of the 62 SNP Panel described herein. The 51 SNP Panel consists of the SNPs shown in FIG. 15.

5 [00105] “HapMap” as used herein refers to a haplotype map of the human genome that is a catalog of common genetic variants that occur in human beings. HapMap describes what these variants are, where they occur in DNA, how they distribute in haplotypes, and their frequencies among people within populations and among populations in different parts of the world. (The International HapMap Consortium. The International HapMap Project. Nature 426, 789-796 (2003)).

10 [00106] “HapMap Analysis” as used herein refers to an analysis that can be conducted using Haploview software (Barrett JC, Fry B, Maller J, Daly MJ. Haploview: analysis and visualization of LD and haplotype maps. Bioinformatics. 2005 Jan 15 [PubMed ID: 15297300]).

15 [00107] “Linkage Disequilibrium” as used herein refers to the non-random association between two or more alleles at two or more loci (not necessarily on the same chromosome) such that certain combinations of alleles are more likely to occur together on a chromosome than other combinations of alleles than would be expected from a random formation of haplotypes from alleles based on their frequencies.

20 A. METHODS OF PREDICTING A CAUCASIAN SUBJECT’S RISK FOR HAVING OR DEVELOPING A COMPLEMENT-MEDIATED DISEASE

[00108] Described herein are methods for determining a Caucasian subject’s susceptibility to having or developing a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more major or minor

25 haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A,

30 H16_51_A, H17_51_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B,

H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. As used herein, “complement thereof” means a SNP, haplotype, or diplotype that is located on the complement DNA or RNA strand of any SNP, haplotype, or diplotype described herein. For example, and not to be limiting, the haplotype sequence “ATGC” can be converted to its complement thereof “TACG”. Therefore, by way of example, if “ATGC” represents a haplotype indicative of a subject’s susceptibility to having or developing a complement-mediated disease, its complement haplotype sequence, “TACG”, also represents a haplotype indicative of a subject’s susceptibility to having or developing a complement-mediated disease. As described herein, the presence of one or more of the haplotypes disclosed herein can indicate a subject’s susceptibility for having or developing a complement-mediated disease. In one aspect, the complement-mediated disease can be age-related macular degeneration (AMD). In another aspect, the complement-mediated disease can include, but is not limited to atypical haemolytic uraemic syndrome (aHUS), membranoproliferative glomerulonephritis type II (MPGN II; also known as dense deposit disease), systemic lupus erythematosus (SLE), pyogenic infections, hemolysis and thrombosis, partial lipodystrophy, hereditary angioedema, paroxysmal nocturnal haemoglobinuria (PNH), rheumatoid arthritis, myocarditis, multiple sclerosis, IgA nephropathy, Henoch–Schoenlein purpura, glaucoma, post-streptococcal disease, anti-phospholipid antibody syndrome, recurrent pregnancy loss, asthma, antibody-mediated cutaneous disease, or anti-nuclear cytoplasmic antigen-associated pauci-immune vasculitis.

[00109] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject has an increased risk of having or developing a complement-mediated disease. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H2_62_A, H2_51_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for determining a Caucasian subject’s susceptibility to having or developing a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype is indicative of the subject’s increased risk for having or developing a complement-

mediated disease. In one aspect, also disclosed are methods of determining a Caucasian subject's increased risk susceptibility to having or developing complement-mediated disease, wherein the method further comprises administering a therapeutic composition to the subject when an increased risk is determined. In one aspect, also disclosed are methods of determining a Caucasian subject's increased risk susceptibility to having or developing complement-mediated disease, wherein the subject has an increased risk of having or developing complement-mediated disease and further comprises administering a therapeutic composition to the subject when an increased risk is determined.

[00110] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject has a decreased risk of having or developing a complement-mediated disease. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H3_62_A, H5_62_A, H11_62_A, H3_51_A, H5_51_A, H10_51_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype is indicative of the subject's decreased risk for having or developing a complement-mediated disease.

[00111] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject does not have an increased risk or a decreased risk of having or developing a complement-mediated disease. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H1_62_A, H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H4_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. Thus, described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated

disease comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype does not indicate that the subject has an increased risk or a decreased risk for having or developing a complement-mediated disease.

5 [00112] As used herein, the term “subject” means an individual. In one aspect, a subject is a mammal such as a human. In another aspect, the subject can be a Caucasian subject. In a further aspect a subject can be a non-human primate. Non-human primates include marmosets, monkeys, chimpanzees, gorillas, orangutans, and gibbons, to name a few. The term “subject” also includes domesticated animals, such
10 as cats, dogs, etc., livestock (for example, cattle (cows), horses, pigs, sheep, goats, etc.), laboratory animals (for example, ferret, chinchilla, mouse, rabbit, rat, gerbil, guinea pig, etc.) and avian species (for example, chickens, turkeys, ducks, pheasants, pigeons, doves, parrots, cockatoos, geese, etc.). Subjects can also include, but are not limited to fish (for example, zebrafish, goldfish, tilapia, salmon, and trout),
15 amphibians and reptiles. As used herein, a “subject” is the same as a “patient,” and the terms can be used interchangeably.

[00113] A haplotype can refer to a set of polymorphisms in a single gene, an intergenic sequence, or in larger sequences including both gene and intergenic sequences, e.g., a collection of genes, or of genes and intergenic sequences. For
20 example, a haplotype can refer to a set of polymorphisms in the CFH-to-F13B locus. The CFH-to-F13B locus comprises the chromosome region of chromosome 1q25-q31 (approximately 196,580,000 to 197,053,000; hg19) that contains the following genes and pseudogenes: complement factor H (CFH) and its truncated isoform (CFHT), the complement factor H related genes 3, 1, 4, 2, and 5 (CFHR3, CFHR1, CFHR4,
25 CFHR2 and CFHR5), a CFHR pseudogene, and factor 13B (F13B). The locus can also include all non-genic DNA, including that extending proximal to CFH and distal to F13B. Blocks of linkage disequilibrium (LD) that initiate within the CFH-to-F13B and extend proximal to CFH and/or distal to F13b are also included in the locus.

[00114] As used herein the term “haplotype” can also refer to a set of single
30 nucleotide polymorphisms (SNPs) found to be statistically associated with each other on a single chromosome. A haplotype can also refer to a combination of

polymorphisms (e.g., SNPs) and other genetic markers (e.g., an insertion or a deletion) found to be statistically associated with each other on a single chromosome.

[00115] The term “polymorphism” refers to the occurrence of one or more genetically determined alternative sequences or alleles in a population. A “polymorphic site” is the locus at which sequence divergence occurs. Polymorphic sites have at least one allele. A diallelic polymorphism has two alleles. A triallelic polymorphism has three alleles. Diploid organisms may be homozygous or heterozygous for allelic forms. A polymorphic site can be as small as one base pair. Examples of polymorphic sites include: restriction fragment length polymorphisms (RFLPs), variable number of tandem repeats (VNTRs), hypervariable regions, minisatellites, dinucleotide repeats, trinucleotide repeats, tetranucleotide repeats, and simple sequence repeats. As used herein, reference to a “polymorphism” can encompass a set of polymorphisms (i.e., a haplotype).

[00116] A “single nucleotide polymorphism (SNP)” can occur at a polymorphic site occupied by a single nucleotide, which is the site of variation between allelic sequences. The site can be preceded by and followed by highly conserved sequences of the allele. A SNP can arise due to substitution of one nucleotide for another at the polymorphic site. Replacement of one purine by another purine or one pyrimidine by another pyrimidine is called a transition. Replacement of a purine by a pyrimidine or vice versa is called a transversion. A synonymous SNP refers to a substitution of one nucleotide for another in the coding region that does not change the amino acid sequence of the encoded polypeptide. A non-synonymous SNP refers to a substitution of one nucleotide for another in the coding region that changes the amino acid sequence of the encoded polypeptide. A SNP may also arise from a deletion or an insertion of a nucleotide or nucleotides relative to a reference allele.

[00117] A “set” of polymorphisms means one or more polymorphism, e.g., at least 1, at least 2, at least 3, at least 4, at least 5, at least 6, or more than 6 polymorphisms known, for example, in the CFH-to-F13B locus.

[00118] As used herein, a “nucleic acid,” “polynucleotide,” or “oligonucleotide” can be a polymeric form of nucleotides of any length, can be DNA or RNA, and can be single- or double-stranded. Nucleic acids can include promoters or other regulatory

sequences. Oligonucleotides can be prepared by synthetic means. Nucleic acids include segments of DNA, or their complements spanning or flanking any one of the polymorphic sites known in the CFH-to-F13B locus. The segments can be between 5 and 100 contiguous bases and can range from a lower limit of 5, 10, 15, 20, or 25
5 nucleotides to an upper limit of 10, 15, 20, 25, 30, 50, or 100 nucleotides (where the upper limit is greater than the lower limit). Nucleic acids between 5-10, 5-20, 10-20, 12-30, 15-30, 10-50, 20-50, or 20-100 bases are common. The polymorphic site can occur within any position of the segment. A reference to the sequence of one strand of a double-stranded nucleic acid defines the complementary sequence and except
10 where otherwise clear from context, a reference to one strand of a nucleic acid also refers to its complement.

[00119] “Nucleotide” as described herein refers to molecules that, when joined, make up the individual structural units of the nucleic acids RNA and DNA. A nucleotide is composed of a nucleobase (nitrogenous base), a five-carbon sugar
15 (either ribose or 2-deoxyribose), and one phosphate group. “Nucleic acids” are polymeric macromolecules made from nucleotide monomers. In DNA, the purine bases are adenine (A) and guanine (G), while the pyrimidines are thymine (T) and cytosine (C). RNA uses uracil (U) in place of thymine (T). As used in the figures, “S” represents short; “L” represents “long, and “D” represents deleted in the context
20 of the SNP analysis. For purposes for the sequence listing “S”, “L” and “D” are represented as “N” which refers to any specific nucleotide in the position identified.

[00120] Exemplary polymorphic sites in the CFH-to-F13B locus are described herein as examples and are not intended to be limiting. These polymorphic sites, SNPs, or haplotypes can also be used in carrying out methods described herein. Moreover, it
25 will be appreciated that these CFH-to-F13B locus polymorphisms are useful for linkage and association studies, genotyping clinical populations, correlation of genotype information to phenotype information, loss of heterozygosity analysis, identification of the source of a cell sample, and can also be useful to target potential therapeutics to cells.

[00121] It will be appreciated that additional polymorphic sites in the CFH-to-F13B locus, which are not explicitly described herein, may further refine this analysis. A
30

SNP analysis using non-synonymous polymorphisms in the CFH-to-F13B locus can be useful to identify variant polypeptides. Other SNPs associated with risk may encode a protein with the same sequence as a protein encoded by a neutral or protective SNP but contain an allele in a promoter or intron, for example, changes the level or site of gene expression. It will also be appreciated that a polymorphism in the CFH-to-F13B locus may be linked to a variation in a neighboring gene. The variation in the neighboring gene may result in a change in expression or form of an encoded protein and have detrimental or protective effects in the carrier.

[00122] As used herein, the term “variant” can also refer to a nucleotide sequence in which the sequence differs from the sequence most prevalent in a population, for example by one nucleotide, in the case of the SNPs described herein. For example, some variations or substitutions in the nucleotide sequence of the CFH-to-F13B locus alter a codon so that a different amino acid is encoded resulting in a variant polypeptide. The term “variant,” can also refer to a polypeptide in which the sequence differs from the sequence most prevalent in a population at a position that does not change the amino acid sequence of the encoded polypeptide (i.e., a conserved change). Variant polypeptides can be encoded by a risk haplotype, encoded by a protective haplotype, or can be encoded by a neutral haplotype. Variant polypeptides can be associated with risk, associated with protection, or can be neutral.

[00123] The methods and materials described herein can be used to determine whether the nucleic acid of a subject (e.g., human) contains a polymorphism, such as a single nucleotide polymorphism (SNP). For example, methods and materials provided herein can be used to determine whether a subject has a variant SNP. Any method can be used to detect a polymorphism in the CFH-to-F13B locus. For example, polymorphisms can be detected by sequencing exons, introns, or untranslated sequences, denaturing high performance liquid chromatography (DHPLC), allele-specific hybridization, allele-specific restriction digests, mutation specific polymerase chain reactions, single-stranded conformational polymorphism detection, and combinations of such methods.

[00124] In one aspect, polymorphisms can be detected in a target nucleic acid isolated from a subject. Typically genomic DNA is analyzed. For assay of genomic

DNA, virtually any biological sample containing genomic DNA or RNA, e.g., nucleated cells, is suitable. For example, in the experiments described in the Examples section herein, genomic DNA was obtained from peripheral blood leukocytes collected from case and control subjects (QIAamp DNA Blood Maxi kit, Qiagen, Valencia, Calif.). Other suitable samples include, but are not limited to, saliva, cheek scrapings, biopsies of retina, kidney or liver or other organs or tissues; skin biopsies; amniotic fluid or CNS samples; and the like. In one aspect RNA or cDNA can be assayed. In one aspect, the assay can detect variant proteins encoded by one or more genes present in the CFH-to-F13B. Methods for purification or partial purification of nucleic acids or proteins from patient samples for use in diagnostic or other assays are known in the art.

[00125] By “isolated nucleic acid” or “purified nucleic acid” is meant DNA that is free of the genes that, in the naturally-occurring genome of the organism from which the DNA of the invention is derived, flank the gene. The term therefore includes, for example, a recombinant DNA which is incorporated into a vector, such as an autonomously replicating plasmid or virus; or incorporated into the genomic DNA of a prokaryote or eukaryote (e.g., a transgene); or which exists as a separate molecule (for example, a cDNA or a genomic or cDNA fragment produced by PCR, restriction endonuclease digestion, or chemical or in vitro synthesis). It also includes a recombinant DNA which is part of a hybrid gene encoding additional polypeptide sequence. The term “isolated nucleic acid” also refers to RNA, e.g., an mRNA molecule that is encoded by an isolated DNA molecule, or that is chemically synthesized, or that is separated or substantially free from at least some cellular components, for example, other types of RNA molecules or polypeptide molecules.

[00126] The identity of bases occupying the polymorphic sites in CFH-to-F13B locus can be determined in a subject, e.g., in a patient being analyzed, using any of several methods known in the art. For example, and not to be limiting use of allele-specific probes, use of allele-specific primers, direct sequence analysis, denaturing gradient gel electrophoresis (DGGE) analysis, single-strand conformation polymorphism (SSCP) analysis, and denaturing high performance liquid chromatography (DHPLC) analysis. Other well known methods to detect polymorphisms in DNA include use of: Molecular Beacons technology (see, e.g.,

Piatek et al., 1998; Nat. Biotechnol. 16:359-63; Tyagi, and Kramer, 1996, Nat. Biotechnology 14:303-308; and Tyagi, et al., 1998, Nat. Biotechnol. 16:49-53), Invader technology (see, e.g., Neri et al., 2000, Advances in Nucleic Acid and Protein Analysis 3826:117-125 and U.S. Pat. No. 6,706,471), nucleic acid sequence based amplification (Nasba) (Compton, 1991), Scorpion technology (Thelwell et al., 2000, Nuc. Acids Res, 28:3752-3761 and Solinas et al., 2001, "Duplex Scorpion primers in SNP analysis and FRET applications" Nuc. Acids Res, 29:20.), restriction fragment length polymorphism (RFLP) analysis, and the like. Additional methods will be apparent to one of skill in the art.

[00127] The term "primer" refers to a single-stranded oligonucleotide capable of acting as a point of initiation of template-directed DNA synthesis under appropriate conditions, in an appropriate buffer and at a suitable temperature. The appropriate length of a primer depends on the intended use of the primer but typically ranges from 15 to 30 nucleotides. A primer sequence need not be exactly complementary to a template but must be sufficiently complementary to hybridize with a template. The term "primer site" refers to the area of the target DNA to which a primer hybridizes. The term "primer pair" means a set of primers including a 5' upstream primer, which hybridizes to the 5' end of the DNA sequence to be amplified and a 3' downstream primer, which hybridizes to the complement of the 3' end of the sequence to be amplified.

[00128] Exemplary hybridization conditions for short probes and primers is about 5 to 12 degrees C, below the calculated T_m . Formulas for calculating T_m are known and include: $T_m = 4^{\circ}\text{C} \times (\text{number of G's and C's in the primer}) + 2^{\circ}\text{C} \times (\text{number of A's and T's in the primer})$ for oligos <14 bases and assumes a reaction is carried out in the presence of 50 mM monovalent cations. For longer oligos, the following formula can be used: $T_m = 64.9^{\circ}\text{C} + 41^{\circ}\text{C} \times (\text{number of G's and C's in the primer} - 16.4)/N$, where N is the length of the primer. Another commonly used formula takes into account the salt concentration of the reaction (Rychlik, supra, Sambrook, supra, Mueller, supra.): $T_m = 81.5^{\circ}\text{C} + 16.6^{\circ}\text{C} \times (\log_{10}[\text{Na}^+][\text{K}^+]) + 0.41^{\circ}\text{C} \times (\% \text{ GC}) - 675/N$, where N is the number of nucleotides in the oligo. The aforementioned formulas provide a starting point for certain applications; however, the design of particular probes and

primers may take into account additional or different factors. Methods for design of probes and primers for use in the methods of the invention are well known in the art.

[00129] The design and use of allele-specific probes for analyzing polymorphisms are described by e.g., Saiki et al., 1986; Dattagupta, EP 235,726, Saiki, WO 89/11548.

5 Briefly, allele-specific probes are designed to hybridize to a segment of target DNA from one individual but not to the corresponding segment from another individual, if the two segments represent different polymorphic forms. Hybridization conditions are chosen that are sufficiently stringent so that a given probe essentially hybridizes to only one of two alleles. Typically, allele-specific probes can be designed to hybridize
10 to a segment of target DNA such that the polymorphic site aligns with a central position of the probe.

[00130] As used herein, “probes” are nucleic acids capable of binding in a base-specific manner to a complementary strand of nucleic acid. Such probes include nucleic acids and peptide nucleic acids (Nielsen et al., 1991). Hybridization may be
15 performed under stringent conditions which are known in the art. For example, see Berger and Kimmel (1987) *Methods In Enzymology*, Vol. 152: *Guide To Molecular Cloning Techniques*, San Diego: Academic Press, Inc.; Sambrook et al. (1989) *Molecular Cloning: A Laboratory Manual*, 2nd Ed., Vols. 1-3, Cold Spring Harbor Laboratory; Sambrook (2001) 3rd Edition; Rychlik, W. and Rhoads, R. E., 1989, *Nucl.*
20 *Acids Res.* 17, 8543; Mueller, P. R. et al. (1993) In: *Current Protocols in Molecular Biology* 15.5, Greene Publishing Associates, Inc. and John Wiley and Sons, New York; and Anderson and Young, *Quantitative Filter Hybridization in Nucleic Acid Hybridization* (1985)). As used herein, the term “probe” includes primers. Probes and primers are sometimes referred to as “oligonucleotides.”

25 [00131] Allele-specific probes can be used in pairs, one member of a pair designed to hybridize to the reference allele of a target sequence and the other member designed to hybridize to the variant allele. Several pairs of probes can be immobilized on the same support for simultaneous analysis of multiple polymorphisms within the same target gene sequence.

30 [00132] The design and use of allele-specific primers for analyzing polymorphisms are described by, e.g., WO 93/22456 and Gibbs, 1989. Briefly, allele-specific primers

are designed to hybridize to a site on target DNA overlapping a polymorphism and to prime DNA amplification according to standard PCR protocols only when the primer exhibits perfect complementarity to the particular allelic form. A single-base mismatch prevents DNA amplification and no detectable PCR product is formed. The method works best when the polymorphic site is at the extreme 3'-end of the primer, because this position is most destabilizing to elongation from the primer.

[00133] In some embodiments, genomic DNA can be used to detect polymorphisms in the CFH-to-F13B locus. Genomic DNA can be extracted from a sample, such as a peripheral blood sample or a tissue sample. Standard methods can be used to extract genomic DNA from a sample, such as phenol extraction. In some cases, genomic DNA can be extracted using a commercially available kit (e.g., from Qiagen, Chatsworth, Calif.; Promega, Madison, Wis.; or Gentra Systems, Minneapolis, Minn.).

[00134] Other methods for detecting polymorphisms can involve amplifying a nucleic acid from a sample obtained from a subject (e.g., amplifying the segments of nucleic acids from the CFH-to-F13B locus using CFH-to-F13B locus-specific primers) and analyzing the amplified nucleic acids. This can be accomplished by standard polymerase chain reaction (PCR, qPCR, & RT-PCR) protocols or other methods known in the art. The amplifying can result in the generation of CFH/F13B allele-specific oligonucleotides, which span the single nucleotide polymorphic sites in the CFH-to-F13B locus. The CFH/F13B specific primer sequences and CFH/F13B allele-specific oligonucleotides can be derived from the coding (exons) or non-coding (promoter, 5' untranslated, introns or 3' untranslated) regions of the CFH-to-F13B locus. In one aspect Genomic DNA from all subjects can be isolated from peripheral blood leukocytes with QIAamp DNA Blood Maxi kits (Qiagen, Valencia, CA). DNA samples can be screened for SNPs in the genes contained within the CFH-to-F13B locus. Genotyping can be performed by TaqMan assays (Applied Biosystems, Foster City, California) using 10 ng of template DNA in a 5uL reaction. The thermal cycling conditions in the 384-well thermocycler (PTC-225, MJ Research) can consist of an initial hold at 95°C for 10 minutes, followed by 40 cycles of a 15-second 95°C denaturation step and a 1-minute 60°C annealing and extension step. Plates can be read in the 7900HT Fast Real-Time PCR System (Applied Biosystems).

5 [00135] Amplification products generated using PCR can be analyzed by the use of denaturing gradient gel electrophoresis (DGGE). Different alleles can be identified based on sequence-dependent melting properties and electrophoretic migration in solution. See Erlich, ed., PCR Technology, Principles and Applications for DNA Amplification, Chapter 7 (W.H. Freeman and Co, New York, 1992).

10 [00136] Alleles of target sequences can be differentiated using single-strand conformation polymorphism (SSCP) analysis. Different alleles can be identified based on sequence- and structure-dependent electrophoretic migration of single stranded PCR products (Orita et al., 1989). Amplified PCR products can be generated according to standard protocols and heated or otherwise denatured to form single stranded products, which may refold or form secondary structures that are partially dependent on base sequence.

15 [00137] Alleles of target sequences can be differentiated using denaturing high performance liquid chromatography (DHPLC) analysis. Different alleles can be identified based on base differences by alteration in chromatographic migration of single stranded PCR products (Frueh and Noyer-Weidner, 2003). Amplified PCR products can be generated according to standard protocols and heated or otherwise denatured to form single stranded products, which may refold or form secondary structures that are partially dependent on the base sequence.

20 [00138] Direct sequence analysis of polymorphisms can be accomplished using DNA sequencing procedures that are well-known in the art. See Sambrook et al., Molecular Cloning, A Laboratory Manual (2nd Ed., CSHP, New York 1989) and Zyskind et al., Recombinant DNA Laboratory Manual (Acad. Press, 1988).

25 [00139] A wide variety of other methods are known in the art for detecting polymorphisms in a biological sample. See, e.g., Ullman et al. "Methods for single nucleotide polymorphism detection" U.S. Pat. No. 6,632,606; Shi, 2002, "Technologies for individual genotyping: detection of genetic polymorphisms in drug targets and disease genes" Am J Pharmacogenomics 2:197-205; Kwok et al., 2003, "Detection of single nucleotide polymorphisms" Curr Issues Biol. 5:43-60).

[00140] Also described herein are methods for determining a female Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the female Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, or a complement thereof.

[00141] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject has an increased risk of having or developing a complement-mediated disease. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H1_62_A, H2_62_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for determining a female Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype is indicative of the female subject's increased risk for having or developing a complement-mediated disease.

[00142] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject has a decreased risk of having or developing a complement-mediated disease. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H3_62_A, H5_62_A, H11_62_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for determining a female Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype is indicative of the female subject's decreased risk for having or developing a complement-mediated disease.

[00143] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject does not have an increased risk or a decreased risk of having or developing a complement-mediated disease. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, or a complement thereof. Thus, described herein are methods for determining a female Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype does not indicate that the female subject has an increased risk or a decreased risk for having or developing a complement-mediated disease.

[00144] Also described herein are methods methods for determining a male Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the male Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, or a complement thereof.

[00145] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject has a increased risk of having or developing a complement-mediated disease. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H2_62_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for determining a male Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype is

indicative of the male subject's increased risk for having or developing a complement-mediated disease.

[00146] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject has a decreased risk of having or developing a complement-mediated disease. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H11_62_A, H3_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for determining a male Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype is indicative of the male subject's decreased risk for having or developing a complement-mediated disease.

[00147] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject does not have an increased risk or a decreased risk of having or developing a complement-mediated disease. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H1_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. Thus, described herein are methods for determining a male Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype does not indicate that the male subject has an increased risk or a decreased risk for having or developing a complement-mediated disease.

[00148] In one aspect, the SNPs, haplotypes, or diplotypes described herein can be determined from a sample obtained from the subject. In another aspect, the subject's SNPs, haplotype, or diplotype can be determined by amplifying or sequencing a nucleic acid sample obtained from the subject. In yet a further aspect, the methods

described herein can further comprise administering a therapeutic composition to the subject or treating the subject with an effective amount of a therapeutic composition.

[00149] As used herein, “treating” refers to the medical management of a patient with the intent to cure, ameliorate, stabilize, or prevent a disease, pathological condition, or disorder. This term includes active treatment, that is, treatment directed specifically toward the improvement of a disease, pathological condition, or disorder, and also includes causal treatment, that is, treatment directed toward removal of the cause of the associated disease, pathological condition, or disorder. In addition, this term includes palliative treatment, that is, treatment designed for the relief of symptoms rather than the curing of the disease, pathological condition, or disorder; preventative treatment, that is, treatment directed to minimizing or partially or completely inhibiting the development of the associated disease, pathological condition, or disorder; and supportive treatment, that is, treatment employed to supplement another specific therapy directed toward the improvement of the associated disease, pathological condition, or disorder. In various aspects, the term covers any treatment of a subject, including a mammal (e.g., a human), and includes: (i) preventing the disease from occurring in a subject that can be predisposed to the disease but has not yet been diagnosed as having it; (ii) inhibiting the disease, i.e., arresting its development; or (iii) relieving the disease, i.e., causing regression of the disease.

[00150] The terms “administering” and “administration” refer to any method of providing a pharmaceutical preparation to a subject. Such methods are well known to those skilled in the art and include, but are not limited to, oral administration, sublingual administration, trans-buccal mucosa administration, transdermal administration, administration by inhalation, nasal administration, topical administration, intravaginal administration, ophthalmic administration, intraaural administration, intracerebral administration, intrathecal administration, rectal administration, intraperitoneal administration, and parenteral administration, including injectable such as intravenous administration, intra-arterial administration, intramuscular administration, intradermal administration, and subcutaneous administration. Ophthalmic administration can include topical administration, subconjunctival administration, sub-Tenon’s administration, epibulbar administration,

retrobulbar administration, intra-orbital administration, and intraocular administration, which includes intra-vitreous administration. Administration can be continuous or intermittent. In various aspects, a preparation can be administered therapeutically; that is, administered to treat an existing disease or condition. In further various aspects, a preparation can be administered prophylactically; that is, administered for prevention of a disease or condition.

[00151] As used herein, the term “therapeutically effective amount” refers to an amount that is sufficient to achieve the desired result or to have an effect on an undesired condition.

[00152] In yet another aspect, the methods for determining a Caucasian subject’s susceptibility to having or developing a complement-mediated disease described herein can further comprise examining the subject with an ophthalmological procedure.

[00153] Various ophthalmological procedures known to persons of ordinary skill in the art can be performed to examine the subject including, but not limited to, autofluorescent imaging techniques, infrared imaging techniques, optical coherence tomography (OCT), Stratus optical coherence tomography (Stratus OCT), Fourier-domain optical coherence tomography (Fd-OCT), two-photon-excited fluorescence (TPEF) imaging, adaptive optics scanning laser ophthalmoscopy (AOSLO), scanning laser ophthalmoscopy, near-infrared imaging combined with spectral domain optical coherence tomography (SD-OCT), color fundus photography, fundus autofluorescence imaging, red-free imaging, fluorescein angiography, indocyanin green angiography, multifocal electroretinography (ERG) recording, microperimetry, color Doppler optical coherence tomography (CDOCT), and visual field assessment. Additionally, the subject can be examined by using the Heidelberg Spectralis, the Zeiss Cirrus, the Topcon 3D OCT 2000, the Optivue RTVue SD-OCT, the Opko OCT SLO, the NIDEK F-10, or the Optopol SOCT Copernicus HR.

1. PREDICTING PROGRESSION OF DISEASE

[00154] Also described herein are methods for predicting progression of a complement-mediated disease in a Caucasian subject. The complement-mediated

disease can be, but is not limited to, age-related macular degeneration. In one aspect the methods comprise determining in the Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof.

As described herein, the presence of one or more of the haplotypes disclosed herein can indicate a subject's susceptibility for having or developing late-stage age-related macular degeneration.

[00155] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject has a increased risk of having or developing late-stage age-related macular degeneration. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H2_62_A, H2_51_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for determining a Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype is indicative of the subject's increased risk for having or developing late-stage age-related macular degeneration.

[00156] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject has a decreased risk of having or developing late-stage age-related macular degeneration. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H3_62_A, H5_62_A, H11_62_A, H3_51_A, H5_51_A, H10_51_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for determining a Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the

Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype is indicative of the subject's decreased risk for having or developing late-stage age-related macular degeneration.

[00157] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject does not have an increased risk or a decreased risk of having or developing late-stage age-related macular degeneration. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H1_62_A, H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H4_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. Thus, described herein are methods for determining a Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype does not indicate that the subject has an increased risk or a decreased risk for having or developing late-stage age-related macular degeneration.

[00158] Also described herein are methods methods for determining a female Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the female Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, or a complement thereof.

[00159] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject has a increased risk

of having or developing late-stage age-related macular degeneration. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H1_62_A, H2_62_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for determining a female Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype is indicative of the female subject's increased risk for having or developing late-stage age-related macular degeneration.

[00160] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject has a decreased risk of having or developing late-stage age-related macular degeneration. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H3_62_A, H5_62_A, H11_62_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for determining a female Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype is indicative of the female subject's decreased risk for having or developing late-stage age-related macular degeneration.

[00161] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject does not have an increased risk or a decreased risk of having or developing late-stage age-related macular degeneration. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, or a complement thereof. Thus, described herein are methods for determining a female Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a

neutral haplotype does not indicate that the female subject has an increased risk or a decreased risk for having or developing late-stage age-related macular degeneration.

[00162] Also described herein are methods for determining a male Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the male Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, or a complement thereof.

[00163] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject has a increased risk of having or developing late-stage age-related macular degeneration. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H2_62_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for determining a male Caucasian subject's susceptibility to having or late-stage age-related macular degeneration comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype is indicative of the male subject's increased risk for having or developing late-stage age-related macular degeneration.

[00164] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject has a decreased risk of having or developing late-stage age-related macular degeneration. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H11_62_A, H3_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for determining a male Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype is indicative

of the male subject's decreased risk for having or developing late-stage age-related macular degeneration.

[00165] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject does not have an increased risk or a decreased risk of having or developing late-stage age-related macular degeneration. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H1_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. Thus, described herein are methods for determining a male Caucasian subject's susceptibility to having or developing late-stage age-related macular degeneration comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype does not indicate that the male subject has an increased risk or a decreased risk for having or developing late-stage age-related macular degeneration.

2. DIPLTYPES

[00166] Also described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more diplotypes described herein. Human autosomal chromosomes normally come in pairs, and the combination in one individual of these two haplotypes is referred to herein as a "Diplotype". As such, "based on" when used in the context of a diplotype "based on" a haplotype, can refer to identifying the combination in one individual of these two haplotypes.

[00167] Any haplotype or haplotypes described herein can be used to create a diplotype. For example, a diplotype can comprise a protective and a risk haplotype, two protective haplotypes, two risk haplotypes, a neutral and a risk haplotype, a neutral and a protective haplotype, or two neutral haplotypes. In an aspect, one haplotype may be dominant or one haplotype may be recessive.

[00168] For example, and not to be limiting, a single H1 haplotype when combined with another H1 haplotype can be an H1_Y_Z:H1_Y_Z diplotype; a single H1 haplotype when combined a H2, H8, or H13 haplotype can be an H1_Y_Z:H2_Y_Z diplotype, an H1_Y_Z:H8_Y_Z diplotype, or an H1_Y_Z:H13_Y_Z diplotype. In one aspect, the diplotypes described herein can be associated with increased risk for developing AMD. In another aspect, the diplotypes described herein can be associated with a decreased risk for developing AMD.

[00169] Described herein are methods for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates the subject's susceptibility for having or developing age-related macular degeneration. In some aspects, the subject's haplotype can be determined from a sample obtained from the subject. In some aspects the subject's haplotype can be determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

[00170] Described herein are methods for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates the subject's susceptibility for having or developing age-related macular degeneration,

[00171] Described herein are methods for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, wherein the diplotype H1_51_B: H1_51_B, H1_51_B:H2_51_B, H1_51_B:H8_51_B, H1_51_B:H13_51_B, H2_51_B:H2_51_B, H2_51_B:H10_51_B, H1_51_B: any minor 51_B haplotype, H2_51_B: any minor 51_B haplotype, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration.

[00172] In some aspects wherein an increased risk is identified, the method can further comprise administering a therapeutic composition to the subject.

[00173] Described herein are methods for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, wherein diplotype H1_51_B: H3_51_B, H1_51_B:H5_51_B, H1_51_B:H14_51_B, H3_51_B: H3_51_B, H3_51_B: H4_51_B, H3_51_B: H5_51_B, H3_51_B: H6_51_B, H3_51_B: any 51_B minor haplotype, H4_51_B: H5_51_B, H5_51_B: H5_51_B, H5_51_B: any 51_B minor haplotype, H6_51_B: H6_51_B, H6_51_B: any 51_B minor haplotype, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

[00174] In some aspects the subject can be female. In some aspects wherein the subject is female, one or more of the diplotypes identified in Figure 37 can be used to indicate the subject's risk for having or developing age-related macular degeneration.

[00175] In some aspects the subject can be male. In some aspects wherein the subject is male, one or more of the diplotypes identified in Figure 36 can be used to indicate the subject's risk for having or developing age-related macular degeneration.

[00176] Described herein are methods of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates whether the subject is in need of treatment for age-related macular degeneration.

[00177] In some aspects, the subject's haplotype can be determined from a sample obtained from the subject. In some aspects the subject's haplotype can be determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

[00178] Described herein are methods of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, wherein diplotype H1_51_B: H1_51_B, H1_51_B:H2_51_B, H1_51_B:H8_51_B, H1_51_B:H13_51_B, H2_51_B:H2_51_B, H2_51_B:H10_51_B, H1_51_B: any minor 51_B haplotype, H2_51_B: any minor 51_B haplotype, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

[00179] Described herein are methods of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B,

H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B,
 H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B,
 H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof,
 , wherein diplotype H1_51_B: H3_51_B, H1_51_B:H5_51_B, H1_51_B:H14_51_B,
 5 H3_51_B: H3_51_B, H3_51_B: H4_51_B, H3_51_B: H5_51_B, H3_51_B:
 H6_51_B, H3_51_B: any 51_B minor haplotype, H4_51_B: H5_51_B, H5_51_B:
 H5_51_B, H5_51_B: any 51_B minor haplotype, H6_51_B: H6_51_B, H6_51_B:
 any 51_B minor haplotype, or a complement thereof, indicates the subject is not in
 need of treatment for age-related macular degeneration.

10 **[00180]** Described herein are methods of identifying a Caucasian subject in need of
 treatment for age-related macular degeneration comprising determining in the
 Caucasian subject the identity of one or more diplotypes based on one or more
 haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B,
 H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B,
 15 H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B,
 H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof,
 wherein the presence of one or more of the diplotypes identified in Figure 36 can be
 used to determine whether, or a complement thereof, can indicates the subject may be
 in need of treatment for age-related macular degeneration. In some aspects the
 20 subject can be female. In some aspects wherein the subject is female, one or more of
 the diplotypes identified in Figure 37 can be used to deterime whether the subject is in
 need of treatment for age-related macular degeneration. In some aspects the subject
 can be male. In some aspects wherein the subject is male, one or more of the
 diplotypes identified in Figure 36 can be used to determine whether the subject is in
 25 need of treatment for age-related macular degeneration.

[00181] Described herein are methods of identifying a Caucasian subject in need of a
 prophylactic treatment for age-related macular degeneration comprising determining
 in the Caucasian subject the identity of one or more diplotypes based on one or more
 haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B,
 30 H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B,
 H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B,
 H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof,

and wherein the presence of one or more of the diplotypes indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

[00182] Described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, wherein diplotype H1_51_B: H1_51_B, H1_51_B:H2_51_B, H1_51_B:H8_51_B, H1_51_B:H13_51_B, H2_51_B:H2_51_B, H2_51_B:H10_51_B, H1_51_B:any minor 51_B haplotype, H2_51_B:any minor 51_B haplotype, or a complement thereof, indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

[00183] Described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, wherein diplotype H1_51_B: H3_51_B, H1_51_B:H5_51_B, H1_51_B:H14_51_B, H3_51_B: H3_51_B, H3_51_B: H4_51_B, H3_51_B: H5_51_B, H3_51_B: H6_51_B, H3_51_B: any 51_B minor haplotype, H4_51_B: H5_51_B, H5_51_B: H5_51_B, H5_51_B: any 51_B minor haplotype, H6_51_B: H6_51_B, H6_51_B: any 51_B minor haplotype, or a complement thereof, indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

[00184] Described herein are methods of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B,

H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B,
 H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B,
 H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof,
 wherein the presence of one or more of the diplotypes identified in Figure 36, or a
 complement thereof, can be used to determine whether, the subject may be in need of
 prophylactic treatment for age-related macular degeneration.

[00185] Described herein are methods of identifying a Caucasian subject appropriate
 for an age-related macular degeneration clinical trial comprising determining in the
 Caucasian subject the identity of one or more diplotypes based on one or more
 haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B,
 H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B,
 H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B,
 H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof,
 and wherein the presence of one or more of the diplotypes indicates whether the
 subject is appropriate for the clinical trial.

[00186] Described herein are methods of identifying a Caucasian subject appropriate
 for an age-related macular degeneration clinical trial comprising determining in the
 Caucasian subject the identity of one or more diplotypes based on one or more
 haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B,
 H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B,
 H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B,
 H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof,
 wherein diplotype H1_51_B: H1_51_B, H1_51_B:H2_51_B, H1_51_B:H8_51_B,
 H1_51_B:H13_51_B, H2_51_B:H2_51_B, H2_51_B:H10_51_B, H1_51_B:any
 minor 51_B haplotype, H2_51_B:any minor 51_B haplotype, or a complement
 thereof, indicates the subject is appropriate for the clinical trial.

[00187] Described herein are methods of identifying a Caucasian subject appropriate
 for an age-related macular degeneration clinical trial comprising determining in the
 Caucasian subject the identity of one or more diplotypes based on one or more
 haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B,
 H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B,

H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, wherein diplotype H1_51_B: H3_51_B, H1_51_B: H5_51_B, H1_51_B: H14_51_B, H3_51_B: H3_51_B, H3_51_B: H4_51_B, H3_51_B: H5_51_B, H3_51_B:

5 H6_51_B, H3_51_B: any 51_B minor haplotype, H4_51_B: H5_51_B, H5_51_B: H5_51_B, H5_51_B: any 51_B minor haplotype, H6_51_B: H6_51_B, H6_51_B: any 51_B minor haplotype, or a complement thereof, indicates the subject is not appropriate for the clinical trial.

10 **[00188]** Described herein are methods of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, 15 H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, wherein the presence of one or more of the diplotypes identified in Figure 36, or a complement thereof, indicates the subject may be appropriate for the clinical trial.

3. HAPLOTYPE TAGGING SNPs

20 **[00189]** Described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can be, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at 25 least six of the SNPs can indicate the subject's susceptibility for having or developing age-related macular degeneration. In another aspect, the SNPs can be haplotype tagging SNPs (htSNPs). As used herein, haplotype tagging SNP means a SNP or multiple SNPs that can identify or represent a haplotype or multiple haplotypes described herein. A tagging SNP or multiple tagging SNPs can be used to identify or 30 depict a haplotype or multiple haplotypes in a subject, and, therefore, can be used to

determine a subject's susceptibility to having or developing a complement-mediated disease.

[00190] In one aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In yet another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In yet another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration.

In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration.

5 **[00191]** Also described herein are methods for determining a female Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the female Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can be, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153,
10 or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs can indicate the subject's susceptibility for having or developing a complement-mediated disease.

[00192] In one aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an G at
15 rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In yet another aspect, an A at
20 rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at
25 rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In yet another aspect, an A at
30 rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In still

another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In sill another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration.

[00193] Also described herein are methods for determining a male Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the male Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can include, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs indicates the subject's susceptibility for having or developing a complement-mediated disease.

[00194] In one aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In yet another aspect, an A at

rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject's increased risk for having or developing age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing age-related macular degeneration.

[00195] Also described herein are methods for predicting progression of a complement-mediated disease in a Caucasian subject comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can be, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs can indicate the subject's risk for having or developing late-stage age-related macular degeneration.

[00196] In one aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In still another aspect, an A at rs35928059, an A at rs800292, a T at

rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In still another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration.

[00197] Also described herein are methods for predicting progression of complement-mediated disease in a female Caucasian subject comprising determining in the female Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can be, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs is indicative of the subject's risk for having or developing late-stage age-related macular degeneration.

[00198] In one aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an G at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration.

rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration.

[00199] Also described herein are methods for predicting progression of a complement-mediated disease in a male Caucasian subject comprising determining in the male Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can include, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs can indicate the subject's risk for having or developing late-stage age-related macular degeneration.

[00200] In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject's increased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153,

or a T at rs698859 can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration.

4. SINGLE SNPs

[00201] Described herein are a collection of polymorphisms and haplotypes comprised of multiple variations in the CFH-to-F13B locus. One or more of these polymorphisms or haplotypes can be associated with complement-mediated disease. Detection of these and other polymorphisms and sets of polymorphisms (e.g., haplotypes) can be useful in designing and performing diagnostic assays for complement-mediated disease. Polymorphisms and sets of polymorphisms can be detected by analysis of nucleic acids, by analysis of polypeptides encoded the CFH-to-F13B locus coding sequences (including polypeptides encoded by splice variants), by analysis of the CFH-to-F13B locus non-coding sequences, or by other means known in the art. Analysis of such polymorphisms and haplotypes can also be useful in designing prophylactic and therapeutic regimes for complement-mediated disease.

[00202] More specifically, described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus. In one aspect, the the SNPs can include, but are not limited to: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In another aspect, the presence of one or more of the SNPs described herein can indicate the subject's susceptibility for having or developing age-related macular degeneration.

[00203] In one aspect, an A at the rs1061170 SNP can be indicative of the subject's increased risk for having or developing a complement-mediated disease. In another aspect, an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP can be indicative of the subject's decreased risk for having or developing age-related macular degeneration. In yet another aspect, a G at the rs3753396 SNP does not indicate that the subject has an increased risk or a decreased risk for having or developing age-related macular degeneration.

[00204] Also described herein are methods for predicting progression of age-related macular degeneration in a Caucasian subject comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus. In one aspect, the SNPs can include, but are not limited to: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In another aspect, the presence of one or more of the SNPs described herein can be indicative of the subject's risk for having or developing late-stage age-related macular degeneration.

[00205] In one aspect, an A at the rs1061170 SNP can be indicative of the subject's increased risk for having or developing late-stage age-related macular degeneration. In another aspect, an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP can be indicative of the subject's decreased risk for having or developing late-stage age-related macular degeneration. In yet another aspect, a G at the rs3753396 SNP does not indicate whether a subject has an increased risk or a decreased risk for having or developing late-stage age-related macular degeneration.

[00206] Also described herein are methods of screening for polymorphic sites in other genes that are in linkage disequilibrium (LD) with a risk, protective, or otherwise informative SNP or genetic marker described herein, including but not limited to the polymorphic sites in the CFH-to-F13B locus. These methods can involve identifying a polymorphic site in a gene that is in linkage disequilibrium with a polymorphic site in the CFH-to-F13B locus, wherein the polymorphic form of the polymorphic site in the CFH-to-F13B locus is associated with complement-mediated disease, (e.g., increased or decreased risk), and determining haplotypes in a population of individuals to indicate whether the linked polymorphic site has a polymorphic form in linkage disequilibrium with the polymorphic form of the CFH/F13B gene that correlates with the complement-mediated disease phenotype. SNPs in LD with the SNPs described herein include, but are not limited to rs10737680, rs7535263, rs10922106, rs395998, rs1329428 and rs7540032. In another aspect, SNPs in LD with the SNPs described herein can be, but are not limited to rs10489456, rs70620, rs742855, rs11799380, rs1065489, rs11582939, rs385390, rs421820, rs426736, rs370953, rs371075.

[00207] As used herein, “linkage disequilibrium” is the non-random association of alleles at two or more loci, not necessarily on the same chromosome. It is not the same as linkage, which describes the association of two or more loci on a chromosome with limited recombination between them. Linkage disequilibrium describes a situation in which some combinations of alleles or genetic markers occur more or less frequently in a population than would be expected from a random formation of haplotypes from alleles based on their frequencies. Non-random associations between polymorphisms at different loci are measured by the degree of linkage disequilibrium (LD). The level of linkage disequilibrium can be influenced by a number of factors including genetic linkage, the rate of recombination, the rate of mutation, random drift, non-random mating, and population structure. “Linkage disequilibrium” or “allelic association” thus means the non-random association of a particular allele or genetic marker with another specific allele or genetic marker more frequently than expected by chance for any particular allele frequency in the population. A marker in linkage disequilibrium with an informative marker, such as one of the CFH/F13B SNPs or haplotypes described herein can be useful in detecting susceptibility to complement-mediated disease. A SNP that is in linkage disequilibrium with a risk, protective, or otherwise informative SNP, haplotype, or genetic marker described herein can be referred to as a “proxy” or “surrogate” SNP. A proxy SNP may be in at least 50%, 60%, or 70% in linkage disequilibrium with risk, protective, or otherwise informative SNP or genetic marker described herein, and in one aspect is at least about 80%, 90%, and in another aspect 91%, 92%, 93%, 94%, 95%, 96%, 97%, 98%, 99%, or about 100% in LD with a risk, protective, or otherwise informative SNP, haplotype, or genetic marker described herein.

[00208] Publicly available databases such as the HapMap database (http://ftp.hapmap.org/ld_data/latest/) and Haploview (Barrett, J. C. et al., Bioinformatics 21, 263 (2005)) may be used to calculate linkage disequilibrium between two SNPs. The frequency of identified alleles in disease versus control populations can be determined using the methods described herein. Statistical analyses can be employed to determine the significance of a non-random association between the two SNPs (e.g., Hardy-Weinberg Equilibrium, Genotype likelihood ratio (genotype p value), Chi Square analysis, Fishers Exact test). A statistically

significant non-random association between the two SNPs indicates that they are in linkage disequilibrium and that one SNP can serve as a proxy for the second SNP.

[00209] In another aspect, polymorphisms in the CFH-to-F13B locus, such as those described herein, can be used to establish physical linkage between a genetic locus associated with a trait of interest and polymorphic markers that are not associated with the trait, but are in physical proximity with the genetic locus responsible for the trait and co-segregate with it. Mapping a genetic locus associated with a trait of interest facilitates cloning the gene(s) responsible for the trait following procedures that are well-known in the art.

[00210] As used herein, “physical linkage” describes the tendency of genes, alleles, loci or genetic markers to be inherited together as a result of their location on the same chromosome. Linkage can be measured by percent recombination between the two genes, alleles, loci or genetic markers. Typically, loci occurring within a 50 centimorgan (cM) distance of each other are linked. Linked markers may occur within the same gene or gene cluster.

5. DELETION TAGGING SNPS

[00211] As disclosed herein, are SNPs that tag the deletion status of the CFHR-3/CFHR-1 genes. Therefore, also described herein are methods for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus. The SNP can be, but is not limited to: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939. In one aspect, the presence of the SNP can indicate the subject's susceptibility for having or developing age-related macular degeneration. In another aspect, a T at the rs12144939 SNP can indicate the subject's decreased risk for having or developing age-related macular degeneration. In another aspect, also described herein are methods determining for determining a Caucasian subject's susceptibility to having or developing a complement-mediated disease comprising determining in the subject the identity of at least one SNP that tags the deletion tagging SNPs described herein. The tagging SNPs can be, but are not limited to, rs6689009, rs35253683, rs731557, and rs60642321. In another aspect, also described herein are methods determining for determining a Caucasian subject's

susceptibility to having or developing a complement-mediated disease comprising determining in the subject the identity of at least one SNP in LD with the deletion tagging SNPs described herein. The SNPs can be, but are not limited to, rs6677604, rs16840522 and rs2019727.

5 **[00212]** Also described herein are methods for predicting progression of a complement-mediated disease in a Caucasian subject comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus. The SNP can include, but is not limited to: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939. In one aspect, the presence of the deletion tagging
10 SNP can indicate the subject's risk for having or developing late-stage age-related macular degeneration. In another aspect, a T at the rs12144939 SNP can indicate the subject's decreased risk for having or developing late-stage age-related macular degeneration.

6. HAPLOTYPE TAGGING HAPLOTYPES

15 **[00213]** In one aspect, the haplotypes described herein can be compared to haplotypes generated using a genetic variation database, for example, HapMap, in order to further refine haplotype association with complement-mediated diseases, such as AMD. The International HapMap Project, available at
20 <http://hapmap.ncbi.nlm.nih.gov/>, contains over 500 SNPs in the region encompassing the CFH-to-F13B locus (Hg18 chr1: 194869137-195303491). Thus, in one aspect, genotypes from Caucasian individuals (referred to as the CEU population) can be downloaded for each of these 500 plus SNPs. Phased genotype data from the CEU set can then be downloaded, including the most informative SNPs (*i.e.* those that segregate in the CEU population) in this region of interest. In one aspect, Haploview
25 (www.broadinstitute.org) can be used to construct haplotypes for the CEU dataset and their frequencies can be calculated. The overlapping SNPs obtained from that analysis can be used as 'tagging' SNPs to match the case/control haplotypes described herein to their equivalent HapMap CEU haplotypes, thereby allowing the disease associations described herein to be extended to the equivalent HapMap haplotypes.

30 **[00214]** In one aspect, the methods can identify HapMap SNPs that were not included in the 51 SNP-based haplotypes described herein. These newly identified

SNPs can further refine the discriminative value of the 51 SNP-based haplotypes described herein, and the association of those haplotypes with complement-mediated disease.

[00215] Described herein are methods for identifying a single nucleotide polymorphism or haplotype that tags one or more haplotypes in the CFH-to-F13B locus. The one or more haplotypes can be any haplotype described herein as being associated with having or developing a complement-mediated disease. In one aspect, the methods can comprise entering the SNP sequence of one or more CFH/F13B haplotypes into a genetic variation database and determining the identity of one or more tagging SNPs or haplotypes. In one aspect, the one or more CFH/F13B haplotypes can be H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof.

[00216] In another aspect, the genetic variation database can be HapMap. In yet another aspect, the one or more CFH/F13B haplotypes can be risk haplotypes, including, but not limited to, H2_62_A, H2_51_A, H1_51_B, H2_51_B, or a complement thereof. In still another aspect, the one or more CFH/F13B haplotypes can be protective haplotypes, including, but not limited to, H3_62_A, H5_62_A, H11_62_A, H3_51_A, H5_51_A, H10_51_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof.

B. METHODS OF IDENTIFYING A CAUCASIAN SUBJECT IN NEED OF TREATMENT FOR AGE-RELATED MACULAR DEGENERATION

[00217] Described herein are methods of identifying a Caucasian subject in need of treatment for complement-mediated disease comprising determining in the Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can

include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A ,
H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A,
H13_62_A, H14_62_A, H15_62_A, H1_51_A, H2_51_A, H3_51_A, H4_51_A,
H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A,
5 H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H1_51_B,
H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B,
H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B,
H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B,
or a complement thereof. As described herein, the presence of one or more of the
10 haplotypes disclosed herein can indicate whether the subject is in need of treatment
for a complement-mediated disease. As used herein, "treatment" can also mean
prophylactic, or preventative treatment. In one aspect, the complement-mediated
disease can be age-related macular degeneration (AMD). Thus, the methods
described herein can be used to identify a Caucasian subject in need of treatment for
15 age-related macular degeneration.

[00218] In one aspect, determining the identity of a certain haplotype or certain
haplotypes in a subject can indicate that the subject is in need of treatment for a
complement-mediated disease. Such haplotypes can be risk haplotypes. For
example, and not to be limiting, a risk haplotype can be H2_62_A, H2_51_A,
20 H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods
for identifying a Caucasian subject in need of treatment for a complement-mediated
disease comprising determining in the Caucasian subject the identity of one or more
haplotypes described herein, wherein a risk haplotype can indicate that the subject is
in need of treatment. In one aspect, the method of identifying a subject in need of
25 treatment can further comprise administering a therapeutic composition to the subject
identified as being in need of treatment. In another aspect, the methods described
herein can be used to identify a Caucasian subject in need of more frequent screening
for complement-mediated diseases, including, but not limited to, age-related macular
degeneration. For example, identifying a subject with a risk haplotype can indicate
30 that the subject needs to be more closely and frequently monitored for the
development of a complement-mediated disease such as AMD.

[00219] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject is not in need of treatment for a complement-mediated disease. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H3_62_A, H5_62_A, H11_62_A, H3_51_A, H5_51_A, H10_51_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype can indicate that the subject is not in need of treatment for a complement-mediated disease.

[00220] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject is in need of continued screening for the development of a complement-mediated disease. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H1_62_A, H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H4_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. Thus, described herein are methods identifying a Caucasian subject in need of screening for the development of a complement-mediated disease comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype can indicate a subject in need of screening for the development of a complement-mediated disease. For example, and not to be limiting, identifying a subject with a neutral haplotype can indicate that the subject should be screened for the development of a complement-mediated disease, such as AMD, at an earlier age, and more often than would typically be done in the general population.

[00221] Also described herein are methods for identifying a female Caucasian subject in need of treatment for complement-mediated disease comprising determining in the female Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to H1_62_A,

H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A,
 H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A,
 H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B,
 H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B,
 5 H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, or a
 complement thereof.

[00222] In one aspect, determining the identity of a certain haplotype or certain
 haplotypes in a female subject can indicate that the female subject is in need of
 treatment for a complement-mediated disease. Such haplotypes can be risk
 10 haplotypes. For example, and not to be limiting, a risk haplotype can be H1_62_A,
 H2_62_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are
 methods of identifying a female Caucasian subject in need of treatment for a
 complement-mediated disease comprising determining in the female Caucasian
 subject the identity of one or more haplotypes described herein, wherein a risk
 15 haplotype can indicate that the subject is in need of treatment. In one aspect, the
 method of identifying a female subject in need of treatment can further comprise
 administering a therapeutic composition to the female subject identified as being in
 need of treatment. In another aspect, the methods described herein can be used to
 identify a female Caucasian subject in need of more frequent screening for
 20 complement-mediated diseases, including, but not limited to, age-related macular
 degeneration. For example, identifying a female subject with a risk haplotype can
 indicate that the female subject needs to be more closely and frequently monitored for
 the development of a complement-mediated disease such as AMD.

[00223] In another aspect, determining the identity of a certain haplotype or certain
 25 haplotypes in a female subject can indicate that the female subject is not in need of
 treatment for complement-mediated disease. Such haplotypes can be protective
 haplotypes. For example, and not to be limiting, a protective haplotype can be
 H3_62_A, H5_62_A, H11_62_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a
 complement thereof. Thus, described herein are methods for identifying a female
 30 Caucasian subject in need of treatment for a complement-mediated disease
 comprising determining in the female Caucasian subject the identity of one or more

haplotypes described herein, wherein a protective haplotype can indicate that the female subject is not in need of treatment for a complement-mediated disease.

[00224] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject is in need of continued screening for the development of a complement-mediated disease. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, or a complement thereof. Thus, described herein are methods identifying a female Caucasian subject in need of screening for the development of a complement-mediated disease comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype can indicate a female subject in need of screening for the development of a complement-mediated disease. For example, and not to be limiting, identifying a female subject with a neutral haplotype can indicate that the female subject should be screened for the development of a complement-mediated disease, such as AMD, at an earlier age, and more often than would typically be done in the general population.

[00225] Also described herein are methods identifying a male Caucasian subject in need of treatment for complement-mediated disease comprising determining in the male Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, or a complement thereof.

[00226] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject is in need of treatment for a complement-mediated disease. Such haplotypes can be risk haplotypes. For

example, and not to be limiting, a risk haplotype can be H2_62_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods of identifying a male Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype can indicate that the male subject is in need of treatment. In one aspect, the method of identifying a male subject in need of treatment can further comprise administering a therapeutic composition to the male subject identified as being in need of treatment. In another aspect, the methods described herein can be used to identify a male Caucasian subject in need of more frequent screening for complement-mediated diseases, including, but not limited to, age-related macular degeneration. For example, identifying a male subject with risk haplotype can indicate that the male subject needs to be more closely and frequently monitored for the development of a complement-mediated disease such as AMD.

[00227] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject is not in need of treatment for complement-mediated disease. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H11_62_A, H3_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for identifying a male Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype can indicate that the male subject is not in need of treatment for a complement-mediated disease.

[00228] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject is in need of continued screening for the development of a complement-mediated disease. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H1_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B,

H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. Thus, described herein are methods for identifying a male Caucasian subject in need of screening for the development of a complement-mediated disease comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype can indicate a male subject in need of screening for the development of a complement-mediated disease. For example, and not to be limiting, identifying a male subject with a neutral haplotype can indicate that the male subject should be screened for the development of a complement-mediated disease, such as AMD, at an earlier age, and more often than would typically be done in the general population.

[00229] Also described herein are methods of identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus. The SNPs can include, but are not limited to: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of one or more of the SNPs can indicate whether the subject is in need of treatment for a complement-mediated disease. In one aspect, an A at the rs1061170 SNP can indicate the subject is in need of treatment for a complement-mediated disease. In another aspect, an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP can indicate the subject is not in need of treatment for a complement-mediated disease. In yet another aspect, a G at the rs3753396 SNP can indicate the subject may be in need of treatment for a complement-mediated disease.

[00230] Also described herein are methods of identifying a Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus. The SNP can include, but is not limited to: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939. In one aspect, the presence of one or more of the SNPs can indicate whether the subject is in need of treatment for age-related macular degeneration. In another aspect, a T at the rs12144939 SNP can indicate the subject is not in need of treatment for age-related macular degeneration.

[00231] Also described herein are methods for identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can include, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs can indicate whether the subject is in need of treatment for age-related macular degeneration.

[00232] In one aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In still another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In still another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153,

or a C at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration.

[00233] Also described herein are methods of identifying a female Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the female Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can be, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs can indicate whether the subject is in need of treatment for age-related macular degeneration.

[00234] In one aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an G at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In still

another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In yet another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration.

[00235] Also described herein are methods of identifying a male Caucasian subject in need of treatment for a complement-mediated disease comprising determining in the male Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can be, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs can indicate whether the subject is in need of treatment for age-related macular degeneration.

[00236] In one aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, a G at rs800292, a C at

rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject is in need of treatment for age-related macular degeneration. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not in need of treatment for age-related macular degeneration.

1. METHODS OF IDENTIFYING PATIENTS FOR CLINICAL TRIALS

[00237] Furthermore, described herein are methods of identifying a Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. As described herein, the presence of one or more of the haplotypes disclosed herein can indicate whether the subject is appropriate for a complement-mediated disease clinical trial. In one aspect, the complement-mediated disease can be age-related macular degeneration

(AMD), and the clinical trial can be an age-related macular degeneration clinical trial. Thus, the methods described herein can be used to identify a Caucasian subject that is appropriate for an age-related macular degeneration clinical trial. As used herein, clinical trial means an experimental study conducted to evaluate the effectiveness and safety of a treatment or medical device by monitoring their effects on a subject or group of subjects. In one aspect, the clinical trial can be conducted to evaluate the effectiveness and safety of medications for a complement-mediated disease, for example, AMD.

[00238] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject is appropriate for a complement-mediated disease clinical trial. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H2_62_A, H2_51_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for identifying a Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype can indicate that the subject is appropriate for the clinical trial. In one aspect, the method of identifying a subject for the clinical trial can further comprise administering a therapeutic composition to the subject identified as being appropriate for the clinical trial.

[00239] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject is not appropriate for a complement-mediated disease clinical trial. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H3_62_A, H5_62_A, H11_62_A, H3_51_A, H5_51_A, H10_51_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for identifying a Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype can indicate that the subject is not appropriate for a complement-mediated disease clinical trial. For example, a subject identified to have a protective haplotype described herein can be excluded from participation in a clinical trial designed to evaluate a treatment for a complement-mediated disease.

[00240] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a subject can indicate that the subject may be appropriate for a complement-mediated disease clinical trial. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be

5 H1_62_A, H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H4_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B,

10 H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. Thus, described herein are methods of identifying a Caucasian subject appropriate for a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype can indicate a subject that may be appropriate for a complement-mediated

15 disease clinical trial.

[00241] Furthermore, described herein are methods of identifying a female Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the female Caucasian subject the identity of one or more major or minor haplotypes. The haplotypes can include, but are not limited to

20 H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B,

25 or a complement thereof. As described herein, the presence of one or more of the haplotypes disclosed herein can indicate whether the female subject is appropriate for a complement-mediated disease clinical trial. In one aspect, the complement-mediated disease can be age-related macular degeneration (AMD), and the clinical trial can be an age-related macular degeneration clinical trial. Thus, the methods

30 described herein can be used to identify a female Caucasian subject that is appropriate for an age-related macular degeneration clinical trial.

[00242] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject is appropriate for a complement-mediated disease clinical trial. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H1_62_A, H2_62_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for identifying a female Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a risk haplotype can indicate that the female subject is appropriate for the clinical trial. In one aspect, the method of identifying a female subject for the clinical trial can further comprise administering a therapeutic composition to the subject identified as being appropriate for the clinical trial.

[00243] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the female subject is not appropriate for a complement-mediated disease clinical trial. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H3_62_A, H5_62_A, H11_62_A, H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for identifying a female Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype can indicate that the female subject is not appropriate for a complement-mediated disease clinical trial. For example, a female subject identified to have a protective haplotype described herein can be excluded from participation in a clinical trial designed to evaluate a treatment for a complement-mediated disease.

[00244] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a female subject can indicate that the subject may be appropriate for a complement-mediated disease clinical trial. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B,

H18_51_B, H19_51_B, H20_51_B, or a complement thereof. Thus, described herein are methods identifying a female Caucasian subject appropriate for a complement-mediated disease clinical trial comprising determining in the female Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral
5 haplotype can indicate a female that may be appropriate for a complement-mediated disease clinical trial.

[00245] Furthermore, described herein are methods of identifying a male Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the male Caucasian subject the identity of one or more major or minor
10 haplotypes. The haplotypes can include, but are not limited to H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B,
15 H16_51_B, H17_51_B, H18_51_B, H19_51_B, or a complement thereof. As described herein, the presence of one or more of the haplotypes disclosed herein can indicate whether the male subject is appropriate for a complement-mediated disease clinical trial. In one aspect, the complement-mediated disease can be age-related macular degeneration (AMD), and the clinical trial can be an age-related macular
20 degeneration clinical trial. Thus, the methods described herein can be used to identify a male Caucasian subject that is appropriate for an age-related macular degeneration clinical trial.

[00246] In one aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject is appropriate for a
25 complement-mediated disease clinical trial. Such haplotypes can be risk haplotypes. For example, and not to be limiting, a risk haplotype can be H2_62_A, H1_51_B, H2_51_B, or a complement thereof. Thus, described herein are methods for identifying a male Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the male Caucasian subject the
30 identity of one or more haplotypes described herein, wherein a risk haplotype can indicate that the male subject is appropriate for the clinical trial. In one aspect, the method of identifying a male subject for the clinical trial can further comprise

administering a therapeutic composition to the male subject identified as being appropriate for the clinical trial.

[00247] In another aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the male subject is not appropriate for a complement-mediated disease clinical trial. Such haplotypes can be protective haplotypes. For example, and not to be limiting, a protective haplotype can be H11_62_A, H3_51_B, H14_51_B, or a complement thereof. Thus, described herein are methods for identifying a male Caucasian subject that is appropriate for a complement-mediated disease clinical trial comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a protective haplotype can indicate that the male subject is not appropriate for a complement-mediated disease clinical trial. For example, a male subject identified to have a protective haplotype described herein can be excluded from participation in a clinical trial designed to evaluate a treatment for a complement-mediated disease.

[00248] In yet another aspect, determining the identity of a certain haplotype or certain haplotypes in a male subject can indicate that the subject may be appropriate for a complement-mediated disease clinical trial. Such haplotypes can be neutral haplotypes. For example, and not to be limiting, a neutral haplotype can be H1_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. Thus, described herein are methods of identifying a male Caucasian subject appropriate for a complement-mediated disease clinical trial comprising determining in the male Caucasian subject the identity of one or more haplotypes described herein, wherein a neutral haplotype can indicate a male that may be appropriate for a complement-mediated disease clinical trial.

[00249] Also described herein are methods of identifying a Caucasian subject appropriate for complement-mediated disease clinical trial comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus. The SNPs

can be, but are not limited to: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of one or more of the SNPs described herein can indicate whether the subject is appropriate for the clinical trial. In one aspect, an A at the rs1061170 SNP can indicate the subject is appropriate for the clinical trial. In another aspect, an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP can indicate the subject is not appropriate for the clinical trial. Thus, for example, when it is determined that a Caucasian subject has an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP, the subject can be excluded from a complement-mediated disease clinical trial. In yet another aspect, a G at the rs3753396 SNP can indicate the subject may be appropriate for the clinical trial.

[00250] Also described herein are methods of identifying a Caucasian subject appropriate for an a complement-mediated disease clinical trial comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus. The SNP can include, but is not limited to: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939. In one aspect, the presence of one or more of the SNPs can indicate whether the subject is appropriate for the clinical trial. In another aspect, a T at the rs12144939 SNP can indicate the subject is not appropriate for the clinical trial.

[00251] Also described herein are methods of identifying a Caucasian subject appropriate for a complement-mediated disease clinical trial comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can be, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs can indicate whether the subject is appropriate for the clinical trial.

[00252] In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject is appropriate for the clinical trial. In yet another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a

G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject is appropriate for the clinical trial. In still another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject is appropriate for the clinical trial. In another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject is not appropriate for the clinical trial. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject is not appropriate for the clinical trial. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject is not appropriate for the clinical trial. In still another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not appropriate for the clinical trial. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not appropriate for the clinical trial. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not appropriate for the clinical trial. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not appropriate for the clinical trial.

[00253] Also described herein are methods of identifying a female Caucasian subject appropriate for a complement-mediated disease clinical trial comprising determining in the female Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can include, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at

least six of the SNPs can indicate whether the subject is appropriate for the clinical trial.

[00254] In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an G at rs698859 can indicate the subject is appropriate for the clinical trial. In yet another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject is appropriate for the clinical trial. In still another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject is appropriate for the clinical trial. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject is appropriate for the clinical trial. In yet another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject is not appropriate for the clinical trial. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 can indicate the subject is not appropriate for the clinical trial. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject is not appropriate for the clinical trial. In yet another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not appropriate for the clinical trial. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not appropriate for the clinical trial. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not appropriate for the clinical trial. In yet another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at

rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not appropriate for the clinical trial.

[00255] Also described herein are methods of identifying a male Caucasian subject appropriate for a complement-mediated disease clinical trial comprising determining in the male Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus. The SNPs can be, but are not limited to: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i). In one aspect, the presence of at least six of the SNPs can indicate whether the subject is appropriate for the clinical trial.

[00256] In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 can indicate the subject is appropriate for the clinical trial. In another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 can indicate the subject is appropriate for the clinical trial. In still another aspect, an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 can indicate the subject is appropriate for the clinical trial. In another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 can indicate the subject is not appropriate for the clinical trial. In yet another aspect, an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 can indicate the subject is not appropriate for the clinical trial. In still another aspect, an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 can indicate the subject is not appropriate for the clinical trial.

[00257] Also described herein are methods of enhancing clinical trials comprising choosing appropriate patient populations for those clinical trials. In one aspect, the methods can be used to ensure patients are identified to participate in clinical trials based upon whether or not they are responsive to the experimental therapeutic agent or agents being studied. The methods can comprise monitoring the condition of

subjects receiving treatment for a complement-mediated disease such as AMD. A successful treatment outcome can be indicated by return of complement pathway activity, such as expression level, biochemical activity (e.g., enzymatic activity of a complement component), or serum auto antibodies against complement pathway associated molecules, from abnormal levels to or toward normal levels. In one aspect, the methods can comprise measuring an initial value for the level of abnormal activity (e.g., abnormal presence of an autoantibody, or abnormal levels of complement pathway molecules) before the subject has received treatment. Repeat measurements can then be made over a period of time. For example, and not to be limiting, that period of time can be about 1 day, 2 days, 5 days, 1 week, 2 weeks, 3 weeks, 1 month, 2 months, 3 months, 4 months, 6 months, 9 months, 1 year, or greater than 1 year. If the initial level is elevated relative to the mean level in a control population, a significant reduction in level in subsequent measurements can indicate a positive treatment outcome. Likewise, if the initial level of a measure marker is reduced relative to the mean in a control population, a significant increase in measured levels relative to the initial level can signal a positive treatment outcome. Subsequently measured levels are considered to have changed significantly relative to initial levels if a subsequent measured level differs by more than one standard deviation from the mean of repeat measurements of the initial level. If monitoring reveals a positive treatment outcome, that indicates a patient that can be chosen to participate in a clinical trial for that particular therapeutic agent or agents. If monitoring reveals a negative treatment outcome, that indicates a patient that should not be chosen to participate in a clinical trial for that particular therapeutic agent or agents.

C. Kits

[00258] Also described herein are kits for utilizing the methods described herein. The kits described herein can comprise an assay or assays for detecting one or more haplotypes in a nucleic acid sample of a subject, wherein the one or more haplotypes are the haplotypes described herein. For example, and not to be limiting, the one or more haplotypes can be H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A,

H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof. In one aspect, the one or more haplotypes can be the haplotype tagging haplotypes described herein as well as the haplotype tagging haplotypes shown in Figure 41.

[00259] The kits described herein can further comprise amplification reagents for amplifying the CHF-to-F13B locus, or any of the genes or nucleic acids described herein.

D. EXAMPLES

[00260] The following examples are put forth so as to provide those of ordinary skill in the art with a complete disclosure and description of how the compounds, compositions, articles, devices and/or methods claimed herein are made and evaluated, and are intended to be purely exemplary of the invention and are not intended to limit the scope of what the inventors regard as their invention. Efforts have been made to ensure accuracy with respect to numbers (e.g., amounts, temperature, etc.), but some errors and deviations should 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.

1. Example 1: Patient Cohorts

[00261] DNA samples derived from well-characterized cohorts of study patients with and without AMD were used for the various studies described herein (see Abrera-Abeleda et al 2006; Allikmets et al 2009; Baird et al 2006; DeAngelis et al 2008; Gold et al 2006; Hageman et al, 2005, 2006, 2011; Lim et al 2012; Robman et al 2010; Wickremasinghe et al 2011; Zhang 2008). These collections consist of over 5,000 DNA samples from Caucasian patients with and without AMD. All cohorts were case-control cohorts, with the exception of two sib-pair cohorts, one highly discordant and one both discordant and concordant (657 subjects in 284 families). Index patients in the sib-pair cohort were aged 50 years or older and had CNV in at

least one eye, as defined by sub-retinal hemorrhage, fibrosis or fluorescein angiographic presence of neovascularization documented at the time or, or prior to, enrolment in the study. The unaffected siblings had normal maculas at an age older than that at which the index patient was first diagnosed with CNV. All individuals in both the case-control and sib-pair cohorts were of European-American descent, over the age of sixty, and matched for age and ethnicity. Patients were examined and photographed by trained ophthalmologists. Urine, serum, plasma, DNA and extensive medical and ophthalmological data were available for the majority of these patients. Stereo fundus photographs were graded according to standardized classification systems as described in the references noted above. A large proportion of the samples in all cohorts were genotyped for variants and SNPs in the CFH to F13B locus, and other genes, as described herein.

[00262] DNA samples from several additional cohorts were also available: 1) early onset drusen cases (EOD) (103); 2) Caucasian newborns (1,870); 3) MPGN II (65); atypical hemolytic uremic syndrome (aHUS) (211); 4) glaucoma (330); 5) Alzheimer (1,214); 6) 45 families from the Utah Genetic Reference Project (UGRP); and 7) individuals from the Human Diversity Panels. Moreover, numerous other systemic and ocular diseases were represented in the collective set of samples.

[00263] Example 2: Determination of Deletion Status and Extended Haplotypes Spanning the Chromosome 1q32 CFH-to-F13B Locus; Discovery Phase

[00264] 2.1 Background

[00265] Recent genetic studies that have shown an association between age-related macular degeneration (AMD) and specific haplotypes of the complement factor H gene (CFH), which lies within the “regulators of complement activation” (RCA) gene cluster on chromosome 1q25-q31 (Hageman et al 2005). Although the understanding of nucleotide polymorphisms in loci associated with AMD has increased, characterization of the disease-associated CFH risk and protective haplotypes has not yet been assessed fully. Recent reports suggest that risk variants of the CFH gene in the Japanese and Chinese populations are either not associated with AMD, differ substantially from the Caucasian population and/or that the risk haplotype occurs at a substantially lower frequency (Lau et al 2006; Gotoh et al 2006).

[00266] Five CFH-related genes (CFHR1, CFHR2, CFHR3, CFHR4 and CFHR5), one CFHR pseudogene (LOC100289145) and one additional gene containing complement SCR domains (Factor 13B, or F13B) lie within the regulators of complement activation (RCA) locus on chromosome 1q32. A large, common deletion that encompasses both the CFHR1 and CFHR3 genes has been identified on one of the CFH protective haplotypes (Hageman et al 2006).

[00267] Due to the strong homology between these seven genes, substantial copy number variation with this 1q32 locus, and the lack of understanding of Complement-Mediated Disease causality within this locus, extended haplotypes encompassing CFH, the five CFHR genes, the CFHR pseudogene (LOC100289145) and Factor 13B (F13B) were determined, and their association with AMD was assessed.

[00268] 2.2 Methods: Study Patient Samples

[00269] DNA samples from study patients, including sib pairs, with and without AMD were used. DNA samples from the 1,073 subjects employed for this study (referred to as '1,073 Cohort', 'GSH Cohort' or 'ParAllele Cohort') were of European-American decent and selected from a larger cohort (see Example 1 above). The average age of subjects was 76.2 years. Fifty-nine percent were female. 56% of subjects had age related maculopathy (ARM) or age-related macular degeneration (AMD) and 34.5% were controls. 0.6% of the patients in this cohort could not be graded.

[00270] 2.3 Methods: Overview of Strategy for CFH-to-F13B Haplotype Determination and Validation in the Discovery Cohort

[00271] Sixty-three SNPs (Figure 2) spanning the CFH-to-F13B region were genotyped in 1,073 Caucasian individuals. Missing genotypes were imputed. The deletion status of the CFHR3/CFHR1 genes was assessed initially using single-strand conformation polymorphism (SSCP) and a qPCR assay developed to detect the deletion on single chromosomes. Genotypes were phased five independent times using PHASE v2.1.1, results were combined, and the most likely phased haplotypes for all 1,073 individuals were extracted. Haplotypes for all chromosomes were subsequently constructed (phased). Haplotype validity was confirmed in three-

generation families. Haplotype and SNP disease associations were assessed using appropriate statistical assessment methods.

[00272] 2.4 Methods: Single Nucleotide Polymorphism (SNPs) Genotyping

[00273] Sixty-three single nucleotide polymorphisms (SNPs) within the CFH-to-F13B locus, including non-genic regions both proximal and distal to the CFH and F13B genes (Figure 2), were genotyped using SSCP-based genotyping [5], Taqman based genotyping (ABI), Sanger sequencing, and molecular inversion probe genotyping (ParAllele BioScience) [6] in 1,073 human genomic DNA samples. SNPs were selected based on significance of association in a prior ParAllele Study (a study in which 1,162 DNA samples were genotyped using the Affymetrix 3K 'Iowa AMD Panel'; the final data yielded successful genotypes for 1,113 samples, with a final number of 3,266 successful assays out 3,308 assays attempted), as well as location to provide relatively dense coverage across the entire CFH-to-F13B locus. Missing values were multiply imputed during haplotype construction using PHASE v. 2.1.1 (Am. J. Hum. Gen. 68:978-989, 2001).

[00274] 2.5 Methods: Identification of CFHR3/CFHR1 Deletion Chromosomes

[00275] The deletion status of the CFHR-3/CFHR-1 genes was assessed using SSCP and a qPCR assay developed to detect the deletion on single chromosomes. SSCP assay was used to identify all individuals homozygous for the CFHR3/CFHR1 deletion. Using a pair of PCR primers that amplify both CFH exon 22 and a homologous region in CFHR1 (see Hageman et al 2006 for primers), one of the resulting SSCP conformations (referred to as 'shift 4') was diagnostic of a subject for which both copies of CFHR1 were absent.

[00276] Real-time quantitative PCR (qPCR) was also used to determine the copy number state of the CFHR3 and CFHR1 genes (Figure 31). A set of five to six independent TaqMan assays measured simultaneously the amount of chromosomal DNA upstream of CFHR3 ('the deletion amplicon') using a 6-FAM fluorescent signal and the endogenous control amplicon of RNase P using a VIC tag (Applied Biosystems RNase P Control Reagent). The sequences of the primers and probe for the deletion assay were designed based on the mrd_3855 amplicon. They were

designated mrd_3855-ANYF (TCT GCT CCT GCT CTT CTT TTC C SEQ ID NO. 285), mrd_3855-ANYR (CTC TAT GCT TGC TAC AAG AGA CAC A SEQ ID NO. 286), and mrd_3855-PROBE (6-FAM-AAC AGC ATG GAA TAT C-MGB SEQ ID NO. 287). The reference amplicon was of RNase P, a single copy gene, and reagents were purchased from Applied Biosystems (part number 4316844) with a VIC fluorescent tag attached. Thus, the FAM tag of the deletion amplicon and the VIC tag of the control amplicon could be detected simultaneously in the same rtPCR well. Applied Biosystems Fast Universal PCR Master Mix provided both the polymerase and ROX exogenous non-amplifying background fluorophore. An Applied Biosystems 7900HT Fast Real-Time PCR System was used for all experiments.

[00277] Following data acquisition, analysis was performed using a program written in the R statistical software language. The optimal fluorescence threshold for detection of the exponential amplification phase was determined using a set of serial dilutions of control samples containing one copy of CFHR3/CFHR1 per chromosome. Uncertainties were propagated at all stages to obtain a distribution of Ct values for all samples and all replicates. A sample was classified as having no deletion chromosomes (homozygous normal) if most sample replicates had Ct values for both the deletion amplicon and RNase P similar to the normal control. Samples heterozygous for the CFHR3/CFHR1 deletion had Ct values for the deletion amplicon approximately one cycle later than expected based on their RNase P Ct values. Finally, samples homozygous for the deletion had either no or very minor amplification of the deletion amplicon. A serial dilution of a normal control was run on each qPCR plate.

[00278] 2.6 Methods: Deletion Prediction

[00279] Deletion status was modeled as a biallelic marker: deletion or no-deletion (normal) for each chromosome. Real-Time qPCR was capable of identifying individuals homozygous (DD) or heterozygous (ND) for the deletion and those not containing a deletion (NN). Based upon the deletion status and the genotyping call of CFH locus SNPs, a decision tree capable of predicting deletion status from genotyped SNPs was created using the rpart library in the R statistical software package. In addition to this predictive tree, the strategy that any individual who was heterozygous

for one of the four SNPs screened in the CFHR3/CFHR1 gene region (rs11807997, rs389897, rs4230, or rs414628) cannot contain a deletion for CFHR3 and CFHR1 was also employed. All of these SNPs are in the central portion of the deletion region and none were present when the CFHR3 and CFHR1 genes were absent.

5 **[00280]** 2.7 Methods: Haplotype Inference

10 **[00281]** The qPCR deletion assay was used to classify 536 (and 801 in a second assay) individuals as homozygous deletion (DD), heterozygous deletion (ND), or homozygous non-deletion (NN). All remaining individuals were classified according to their genotypes for SNPs in CFHR3 and CFHR1. All individuals containing more than one heterozygous SNP in CFHR3 or CFHR1 were assumed to be not consistent with a deletion on either chromosome (genotype NN). All other individuals were assigned to have one normal chromosome and one unknown chromosome (N?). All heterozygous deletion individuals were assigned one unknown allele for those SNPs between CFH and CFHR4, the deleted region; homozygous SNPs in this region (ex. 15 CC) were replaced with one known and one unknown allele (C?) while heterozygous SNPs, being inconsistent with a heterozygous deletion, were replaced with two unknown alleles (??). All homozygous deletion individuals were assigned two unknown alleles (??) for each SNPs in the same region.

20 **[00282]** PHASE version 2.1.1 was used to impute missing genotypes and phase genotypes for each individual, care was taken to correctly handle the presence or deletion of CFHR3/1 SNPs as a group. Chromosomal positions for each SNP were obtained from NCBI dbSNP build 129 using the reference human genome (build 18). All individuals typed at SNPs in the CFHR3/CFHR1 region and seven distinct four-SNP haplotypes were imputed for the CFHR3/CFHR1 deletion region using the four 25 SNPs in the deletion region spanning from rs11807997 through rs414628. Five independent runs of PHASE were used to confirm haplotype consistency and two haplotypes were assigned to each individual. Seven distinct four-SNP haplotypes were imputed for this region. Although the first two haplotypes were sufficient to phase 99% of the chromosomes, all seven imputed haplotypes were utilized later to 30 avoid over-fitting of the phased genotypes.

[00283] Deletion status was subsequently treated as an octa-allelic SNP – one for each of the seven imputed CFHR-3/CFHR-1 haplotypes and an eighth allele to signify the deletion. The entire CFH locus was thus modeled as 58 bi-allelic and one octa-allelic SNP. Five independent runs of PHASE were used to impute and phase the genotypes. Five independent runs of PHASE were used to confirm phased genotype consistency. Following genotype phasing, the multi-allelic deletion status was expanded back into four separate tri-allelic SNPs, with a D allele representing the deletion haplotype. In some assessments, all minor haplotypes with frequency less than 1% were iteratively re-assigned to the most similar remaining haplotype.

[00284] PHASE provided probability-weighted, imputed, and phased genotypes for all individuals. Each individual run of PHASE produced multiple weighted estimates of each individual's phased genotypes (using the pairs output file from PHASE). The five independent runs of PHASE produced up to five different imputations of missing genotypes, all of which were then equally weighted. To assign a final pair of haplotypes to each individual, the following algorithm was employed: 1) Input was a collection of individuals where each individual had a set of potential diplotypes and its estimated probability (from PHASE pairs output files); 2) The active set was initialized to include all possible haplotypes; 3) Individual diplotypes were assigned based on the individual's diplotype probabilities with the constraint that diplotypes could only use haplotypes in the active set. It was possible for an individual to not be assigned a diplotype if all of the individual's potential diplotypes required haplotypes not in the active set; 4) Step #7 was skipped to if any individual was not assigned a diplotype; 5) The assigned individual diplotypes were saved as the current solution; 6) The least-used haplotype was removed from the active set and step #3 was returned to; 7) The current solution was outputted as the list of individual diplotypes, and thus haplotypes, present in the population.

[00285] Phased genotypes were also processed in some analyses using Beagle (version 3.0.1) to obtain two haplotype networks — one merging similar haplotypes in the 5' → 3' direction, the second in the 3' → 5' direction. All ten phased genotypes for each individual from the five independent runs of PHASE were used as input to account for uncertainty in the phasing or imputation of missing genotypes. The scale parameter for Beagle's haplotype merging process was set to 4.0. The directed

acyclic graph haplotype output by Beagle indicated which SNPs form haplotype blocks and how those blocks were interrelated. Each chromosome was assigned a haplotype based on its path through the two networks leading to the rs4230 SNP. Haplotypes accounting for less than 1% of the chromosomes were not reported.

5 **[00286]** 2.8 Methods: Haplotype Validation

10 **[00287]** Reference control phased haplotypes were obtained from the members of the Utah Genetic Reference Project families (Ann. Rev. Genomics Hum. Genet. 9:347-358 2008). The UGRP family members consisted of over 600 individuals from 43 three-generation families that were also part of the original Utah CEPH (the Center d'Etude du Polymorphisme Humain) families. Sixty-six SNP genotypes were obtained on blood DNA samples from the entire UGRP members using the Affymetrix Genome-Wide Human SNP array 6.0. Genotype calling was carried out using the CRLMM calling algorithm from the Bioconductor 2.3 Oligo Package. Genotype calls whose confidence was less than 0.95 were replaced with missing values. SNP markers whose call rate was less than 95% were also dropped. Taqman SNP genotyping assays were also carried out to obtain genotypes for 23 additional SNP markers. PedCheck program was used to detect Mendelian inconsistencies and misinherited genotype calls were removed. Phased haplotypes were constructed based on the Mendelian inheritance model in each of the three-generation families. None of the haplotype inference programs were used. Haplotype frequencies were calculated on 136 unrelated UGRP individuals, who were mostly first generation grandparents, using Haploview program's phased haplotype input mode, Haps format (Figure 33).

15 **[00288]** 2.9 Methods: AMD Disease Association

25 **[00289]** Association of haplotypes with AMD was assessed by Bayesian logistic regression in individual SNPs, in 536 individuals (median age = 79 yrs): 216 individuals without AMD and 320 individuals with AREDS categories 2, 3, geographic atrophy, or neovascular AMD. Complement-Mediated Disease association was determined using a Bayesian logistic regression model with Cauchy priors on the coefficients (Ann. Appl. Stat. 2:1360-1383, 2008). Single SNP associations in this case-control cohort were also assessed.

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[00290] 2.10 Results: Genotyping and Identification of Novel CFH Locus SNPs

[00291] DNA from each subject was screened for 41 to 63 of the 63 SNPs employed (Figure 2 and Figure 29). Of the SNPs genotyped by ParAllele using MIP genotyping, five were unreported in the literature (Figure 27) at the time of their discovery. These were identified using mismatch repair detection (MRD) to amplify a variant-enriched pool of amplified genomic DNA. This pool of amplified DNA was then sequenced to identify rare alleles. The rare mrd_3876 and mrd_3877 SNPs were located within the promoter region of CFHR4. The minor allele of mrd_3876 was present in haplotype(s) that skewed toward risk for AMD, and the minor allele of mrd_3877 occurred in haplotype(s) that skewed significantly with AMD risk. The minor allele of mrd_3902, located within exon 2 of CFHR2, results in a Cys72Tyr coding mutation and occurred in haplotypes that appeared to skew toward protection for AMD. The minor alleles of SNPs mrd_3905 and mrd_3906 were more common (minor allele frequencies of approximately 20%) and were located within the promoter region of CFHR5; they did not appear to skew with AMD risk or protection (see Figures 1 and 5).

[00292] One additional novel SNP was identified within the CFH-to-F13B locus at the time these assays were conducted. This variant is located within CFH exon 10A – the last exon of the truncated form of complement factor H (Figure 2). This SNP was identified via SSCP and from sequencing is thought to be a one or two base-pair insertion into the 3' untranslated region of exon 10A. The minor allele, the 1-2 bp deletion, was present in only 1.2% of chromosomes.

[00293] 2.11 Results: Deletion Chromosome Identification - TaqMan qPCR Copy Number Variation Assay

[00294] The results of the deletion assay are shown in FIG. 28. Homozygous deletion samples were verified by SSCP analysis. Of those samples that gave definitive copy number values, 28% had a deletion of one or both copies of CFHR3 and CFHR1. 15% of individual chromosomes did not contain these genes. After including imputed deletion status, 19% of individual chromosomes did not contain the deletion (absence of CFHR3 and CFHR1).

[00295] Experiments were also conducted to determine the reliability of this assay. As an example, one experiment produced 192 replicates for each of two DNA samples -- one homozygous non-deletion sample and a related heterozygous deletion sample. Each qPCR assay was performed using total genomic DNA between 100ng and 0.6ng. Based on these data, for a given amount of genomic DNA, an assay using heterozygous deletion DNA was predicted to have a Ct value 0.645 ± 0.002 cycles greater than an identical assay utilizing homozygous normal DNA. The expected distance from the line $Ct_{\text{deletion}} = Ct_{\text{RNaseP}}$ for an individual sample was 0.16 cycles. Thus, the expected power for correctly classifying a heterozygous deletion of the CFHR3/CFHR1 genes using five to six qPCR replicates was 83%. This is similar to the 75% call rate shown in (Figure 28). Thus, to obtain a power of 95% with a 5% type-I error rate, nine replicates were run on each DNA sample.

[00296] 2.12 Results: Deletion Prediction

[00297] Based upon measured (not imputed) genotypes, a decision tree (Figure 30) was made to predict deletion status from three SNPs. All homozygous deletion individuals had a TT genotype at SNP rs12144939 in CFH, although 17 heterozygous deletion individuals had genotype GG at this location. Thus, it was determined that deletion status in Caucasians can be predicted reliably by a 'T' at rs12144939 located within intron 15. The deletion status of the CFHR3/CFHR1 genes could be predicted with 97% specificity and 96% sensitivity based on the genotype of this single SNP. Fourteen percent of chromosomes in the discovery cohort contained this deletion. The inclusion of rs12406509 and rs7546940 (mrd_3878) slightly increased the sensitivity and specificity of the deletion prediction.

[00298] 2.13 Results: Identification and Validation of 63 SNP-based CFH-to-F13B Haplotypes

[00299] 63 SNP-based haplotypes (Figure 32) spanning the extended CFH-to-F13B locus and including the CFHR3/CFHR1 deletion were determined. Nineteen major haplotypes (>1% frequencies) were identified within the approximately 260kb CFH-to-F13B locus based on haplotype construction using PHASE v.2.1.1 (Figure 32). Haplotype fidelity was confirmed in large, multi-generation Utah Genetic Reference Project families (Annu. Rev. Genomics Hum. Genet. 9:347-358 2008) (Figure 33).

Haplotypes H1 and H2, the two most prevalent haplotypes (approximately 47% of the chromosomes in this cohort), contained the vast majority of the CFH Y402H variant. Five of the 19 haplotypes (H3, H8, H10, H14, and H19) contained the vast majority of all chromosomes with CFHR3/CFHR1 deletions. The CFH I62V protective variant was contained primarily in haplotypes H4, H9, and H12. Eleven SNPs (arrows in **Figure 32**) were sufficient to infer the nineteen haplotypes.

[00300] 2.15 Results: Association of 63 SNP-based CFH-to-F13B Haplotypes with Disease

[00301] Preliminary assessment of AMD disease association showed significant associations of three of the nineteen major haplotypes with different AMD risk phenotypes (as a binary and quantitative trait) ($p < 0.01$), after computing bootstrap confidence intervals and controlling for age, sex, and smoking status. After controlling for age, gender, and smoking status, haplotypes H3, H4, and H8 were associated with significant decreased risk of developing AMD both as binary ($p < 0.01$) and as a quantitative trait ($p < 0.05$) (Figure 34).

[00302] Example 3: Refined Assessment of Extended Haplotypes Spanning the Chromosome 1q32 CFH-to-F13B Locus

[00303] 3.1 Methods: Study Cohorts

[00304] The same 1,073 subject cohort ('1,073 Cohort', 'GSH Cohort' or 'ParAllele Cohort') described in Example 2 was employed in studies to refine the understanding of extended CFH-to-F13B haplotypes and their association with AMD and other diseases. In addition, the highly discordant sib-pair cohort (657 subjects from 284 families), a combined AMD case-control cohort comprised of subjects ascertained in Utah (837 subjects), Iowa (1,555 subjects), Australia Melbourne (1,704 subjects) (this collective cohort is referred to as the 'Combined Cohort') and the UGRP cohort (604 subjects) were employed.

[00305] 3.2 Methods: Genotyping

[00306] 62 SNPs of the 63 SNPs employed in Example 2 were employed in the refined assessment. It was determined that one of the SNPs used to generate the 63

SNP haplotypes, rs432366, was not polymorphic in the location for which it was listed in the NCBI and other databases. Thus, this SNP was removed from all future analyses. Haplotype determinations and disease associations were reassessed based on the removal of rs432366. Genotype call rates exceeding 99% were obtained for 51 of the 62 variants and less than 99% for the remaining SNPs; missing genotypes were imputed using PHASE 2.1.1. Genotyping on all other cohorts was performed using either pre-designed or custom ordered Taqman assays (ABI) and, as such, limited to 51 SNPs (see bolded SNPs in Figure 2 and Figure 16).

[00307] 3.3 Methods: Generation of 62 SNP-based and 51 SNP-based Haplotypes

[00308] PHASE 2.1.1 was employed to determine the SNP-based haplotypes present in the 1,073 patient cohort. Chromosomal positions used were from the NCBI dbSNP build 129 using the reference human genome (build 18). Following genotype phasing, the multi-allelic deletion status was expanded back into four separate tri-allelic SNPs (major, minor, and deleted).

[00309] In addition, 51 SNP-based haplotypes were generated for the highly discordant sib-pair cohort, the AMD case-control cohorts ascertained in Utah, Iowa, Australia ('Combined Cohort') and the UGRP cohort. Of the 62 SNPs employed for the 62 SNP-based haplotype determinations, the four SNPs lying within the CFHR3/CFHR1 deletion region in the 62 SNP-based set (rs11807997, rs389897, rs4230 and rs14628) were not used (instead, the deletion tagging SNP rs12144939 was used as a proxy) for generating the 51 SNP-based haplotypes. In addition, Taqman assays could not be designed to genotype 7 of the remaining 58 SNPs, so these were also not used for generating the 51 SNP-based haplotypes (see Figures 15 and 16 for SNPs employed to generate 51 SNP-based CFH-to-F13B haplotypes).

[00310] 3.4 Methods: Validation of 51 SNP-based Haplotypes

[00311] To validate haplotypes and their frequencies, reference control phased haplotypes were obtained from the members of the Utah Genetic Reference Project (UGRP) families (Annu. Rev. Genomics Hum. Genet. 9:347–358, 2008). TaqMan SNP genotyping assays were used to obtain genotypes for the same 51 additional SNP markers previously genotyped in the GSH cohort (see above). PedCheck program

was used to detect Mendelian inconsistencies and misinherited genotype calls were removed. Phased haplotypes were constructed based solely on the Mendelian inheritance model in each of the three-generation families. None of the haplotype inference programs were used. Haplotype frequencies were calculated on 136
5 unrelated UGRP individuals, who were mostly first generation grandparents, using Haploview program's phased haplotype input mode, Haps Format (Bioinformatics 21:263-265, 2005).

[00312] 3.5 Methods: Disease Association Assessment Using Single Markers and 62
SNP-based and 51 SNP-based Haplotypes

10 [00313] For determination of unrelated case/control disease (AMD) associations using 62 SNP-based haplotypes, only subjects 60 years or older in the 1,073 Cohort were used. Controls were defined as subjects with AMD grades 0, while cases consisted of all subjects with grades 1B-4C (as described in Example 1). Using these
15 criteria, 927 samples from the GSH Cohort (the '927 GSH Cohort') were employed for analysis of disease association. Frequencies of extended haplotypes in the GSH 927 cohort used for disease association were nearly identical to those imputed for the entire 1,073 GSH Cohort (Figure 4). AMD disease association with the 62 SNP-based haplotypes was assessed using the Vassar 2x2 contingency table for Chi-2 based on allele frequencies.

20 [00314] 51 SNPs were also genotyped in the Extremely Discordant Sib-pair Cohort (657 subjects from 284 families). Haploview (v4.2) was used to determine descriptive statistics using normal subjects (only one normal sibling per family for the Family Cohort), *i.e.* deviation from Hardy Weinberg Equilibrium (HWE) and the minor allele in the population. Tests for association of the minor allele with AMD
25 were performed assuming an additive genetic model, and all AMD subtypes were compared to normals in the entire cohort, and then in males only and separately females only. For the Family Cohort, tests for association in the entire cohort were performed using Conditional Logistic Regression (CLR). In the analysis of males and females only, one subject of each gender was chosen per family (choosing the subject
30 with the most severe disease), and tested for association with all AMD subtypes using logistic regression. Logistic regression was also used to assess 51 SNP-based

haplotypes derived from the '927 GSH Cohort'. All analyses were performed in SAS (v9.1) and/or Haploview. Meta-Analysis was performed using Comprehensive Meta-Analysis 2.0.

[00315] 3.6 Results: 62 SNP-based CFH-to-F13B Haplotypes Generated from the 1,073 Cohort

[00316] To generate haplotypes encompassing the CFH-to-F13B locus, 1,073 unrelated Caucasians of Northwestern European decent, were genotyped for 62 SNPs using a combination of assays, with missing genotypes imputed using PHASE2.1.1, as described above. Haplotypes for all samples were then determined using PHASE 2.1.1.

[00317] This analysis resulted in detection of 15 major haplotypes (H1_62_A through H15_62_A; Figure 1 and Figure 17) present at frequencies greater than 1% and 148 minor haplotypes, each present at less than 1% frequency (H16_62_A through H163_62_A; Figure 3 and Figure 17). In total, major haplotypes accounted for 1,599 total chromosomes, while the minor haplotypes contained 547 chromosomes, or 25.5% of the chromosomes in the cohort. The hierarchical relationships between the 163 62 SNP-based haplotypes is shown in Figure 21 (H1_62_A through H163_62_A). The 163 haplotypes were clustered based on the number of similar loci into this tree using the R statistical package hclust function using hierarchical clustering analysis. Similar haplotypes are the most related (closer together) and dissimilar haplotypes are further apart in this 'tree', shown in Figure 21. Importantly, three major clusters containing risk, protective and neutral haplotypes, respectively, were observed. The neutral haplotypes clustered at the left, or top, portion of the tree. Specifically, the neutral haplotype cluster spanned the region from H126_62_A to H163_62_A. The risk haplotypes clustered in the middle portion of the tree. Specifically, the risk haplotype cluster spanned the region from H46_62_A to H73_62_A. The protective haplotypes clustered at the right, or bottom, portion of the tree. Specifically, the protective haplotype cluster spanned the region from H152_62_A to H98_62_A.

[00318] The two most frequent of the 15 major haplotypes, H1_62_A (19.2% of total haplotypes) and H2_62_A (15.4% of total haplotypes) both contained the risk allele

“C” for rs1061170 (402H). H1_62_A and H2_62_A were identical from the region proximal to CFH through CFHR2, based on the variants examined within this region; only in the F13B gene (beginning with rs698859 in the 3’ UTR of F13B), do SNPs distinguish the H1_62_A and H2_62_A haplotypes (Figure 1). Other F13B SNPs that distinguish H1_62_A and H2_62_A include rs5998, rs2990510 and rs1615413. Three other major haplotypes contained the CFH 402H risk allele (H8, H12, H14). Two major haplotypes (H3_62_A and H6_62_A) contained the protective minor allele “A” for CFH rs800292. Additionally, 4 major haplotypes contained the CFHR3/CFHR1 deletion (H5_62_A, H11_62_A, H13_62_A, and H15_62_A; Figure 1). Four SNPs in near perfect LD (rs11807997, rs389897, rs4230, and rs414628) were located in the CFHR3/CFHR1 deletion region; they are shown as tri-allelic SNPs in Figure 1 (major allele shown in white, minor allele shown in line-filled boxes, and deleted CFHR3/CFHR1 shown in darkly shaded boxes).

[00319] Extensive assessment of the 1,073 cohort (see Example 2), as well as families from the UGRP cohort, resulted in the identification of rs12144939 as a reliable surrogate “tag” for the CFHR3/CFHR1 deletion in >98% of Caucasian individuals, with the minor allele “T” accurately identifying an accompanying deletion. Surrogate proxies for this SNP that were in complete LD include rs6677604, rs16840522 and rs2019727. Major haplotypes containing the deletion of CFHR3/CFHR1 (H5_62_A, H11_62_A, H13_62_A and H15_62_A; Figure 1 and Figure 18), with the exception of H13_62_A, are highly protective against the development of AMD in this cohort (Figure 18).

[00320] Importantly, all major haplotypes containing either the protective allele at rs800292 (H5_62_A, H11_62_A, H13_62_A, and H15_62_A) or the CFHR3/CFHR1 deletion (H5_62_A, H11_62_A, H13_62_A, and H15_62_A; include rs11807997, rs389897, rs4230 and rs414628 and are tagged by rs12144939 or surrogate SNPs, as listed above) also contain the minor alleles of the rs2274700, rs3753395, rs393955, rs403846, and rs1410996 variants (Figure 1 and Figure 18). The variant rs1409153 is also in near complete LD with all major haplotypes associated with protection. Any of these SNPs, and any other SNP in near LD with these SNPs, singly or in combination, can serve as surrogates for the major protective haplotypes H3_62_A, H6_62_A, H5_62_A, H11_62_A, H13_62_A, and H15_62_A, as well as some of the

minor protective haplotypes. Other SNPs within this region of DNA that are in near complete LD (and can serve as surrogates) were determined by generating an LD plot using all of the SNPs in HapMap lying within the CFH-to-CFHR4 region. This was done using the CEU cohort (CEU, Utah residents with Northern and Western European Ancestry from the CEPH Collection, is one of the 11 populations in HapMap Phase 3) genotypes. In this population, 7 SNPs were identified to be in complete or near complete LD with rs1410996, the most significantly associated variant in this “block.” They were (in order of 5’-to-3’ direction) rs10737680, rs7535263 (LD = 0.96), rs3753395, rs10922106, rs395998, rs1329428 and rs7540032. There are multiple other SNPs in this 22kb region that are in various levels of LD with rs1410996.

[00321] 3.7 Results: 51 SNP-based CFH-to-F13B Haplotypes in the 1,073 (927 Subset) Cohort

[00322] 51 SNP-based haplotypes were also generated using the 1,073 (927 Subset) Cohort (Figure 22). Converting from 62 to 51 SNPs caused some of the 62 SNP-based haplotypes to collapse into the 51 SNP-based haplotypes (Figure 20). 17 major haplotypes occurred at frequencies greater than 1% in this cohort.

[00323] 3.8 Results: Validation of 62 SNP-based and 51 SNP-based Haplotypes Using the UGRP Cohort

[00324] Because haplotypes were imputed, independent verification was sought that the haplotype structures and frequencies determined were real and representative of a Caucasian population. 62 SNP-based haplotypes were validated as described in see Example 2.

[00325] Using 136 unrelated individuals from the CEPH/UGRP reference cohort, non-imputed haplotypes were generated using 51 SNPs. Given the small size of the UGRP unrelated cohort, some differences in haplotype frequency between the GSH Cohort and the UGRP cohort was expected. However, when compared to the haplotypes shown in Figure 1, these differences were minor and haplotype frequencies were nearly similar (Figure 33), indicating that these haplotype frequencies were representative of aged Caucasian populations in general (Figure 10).

[00326] 3.9 Results: Single SNP AMD Disease Associations in the 1,073 Cohort (927 Subset) Using 62 SNPs

[00327] Of the 1,073 samples in our original cohort, 927 were 60 years or older at diagnosis and had AMD (Grade 1B-4C) or did not have AMD and served as controls (Grade 0). A case/control analysis of risk was performed using logistic regression for single marker associations (**Figure 5**). Results showed patterns of association. rs1061170 was significantly associated with risk ($p=2.5e-11$, OR=1.953, 95% CI 1.604-2.377). The minor alleles of rs1410996, rs2274700, and rs3753395 [and other surrogates tagging the block of DNA containing variants that associate with the majority of protective haplotypes (chr1:196679455-196701284; Hg19)], were even more strongly associated than rs1061170, but with protection from developing, rather than risk of developing AMD ($p=1.12e-14$, OR=0.436, 95% CI 0.353-0.538). Variants in the distal region of this locus including CFHR5 and F13B were also significantly associated with AMD, albeit at much lower levels (Figure 5).

[00328] 3.10 Results: 62 SNP-based Haplotypes Associated with AMD Risk: 1,073 Cohort (927 Subset)

[00329] Disease associations were analyzed for the entire extended haplotypes using Haploview 4.2 (Figure 6) (Barrett JC, Bioinformatics, 2005 Jan 15).

[00330] Of the 15 major haplotypes present at a frequency greater than 1%, H2_62_A exhibited significant risk for AMD (Figure 6 and Figure 18; $p<0.0001$; note that values vary slightly between Figures 6 & 18 due to clinical reclassification of a few patients between the time these two analyses were conducted). H1_62_A, which also contains the 402H risk allele trended slightly toward risk for AMD ($p=0.098$) in this cohort, but was less significant than H1_62_A. Whereas four distal SNPs from the 62 SNP panel distinguish H1_62_A from H2_62_A, only one of these SNPs significantly associated with AMD in single marker assessment (rs698859, $p=0.0111$) (Figure 5). This indicated that additional features/elements in the haplotypes tagged by these SNPs are responsible either for risk in H2_62_A or lack of risk in H1_62_A. Given multiple haplotypes contain the risk allele at rs1061170 (H1_62_A, H2_62_A, H8_62_A, H12_62_A, and H14_62_A) and yet only one of these haplotypes associated significantly with risk in this cohort, there is likely a risk factor present in

the H2_62_A haplotype that is lacking in all other non-H2_62_A 402H haplotypes. In fact, previous studies using less extended haplotypes that failed to include the F13B SNPs have attributed a significant risk of developing AMD to all patients containing the Y402H risk allele (42.3% of major haplotypes or 49% of all haplotypes in the GSH cohort), when in fact risk only associated with H2_62_A, or 16.5% of the cohort described herein. Thus, the haplotypes described herein represent a significant increase in the sensitivity of defining risk at the CFH locus.

[00331] Furthermore, the vast majority of AMD risk association, although not all, occurred in major haplotypes tagged by CFH 402H (H2_62_A, H8_62_A, H12_62_A, H14_62_A and H16_62_A; Figure 6 and Figure 18) in this cohort. CFH 402H, however, was not always associated with risk for AMD; H2_62_A was the most strongly associated haplotype with AMD risk, whereas major haplotypes H1_62_A, H8_62_A, H12_62_A, and H14_62_A were not significantly associated in this cohort. The minor haplotype H16_62_A trended toward risk in this cohort.

[00332] 3.11 Results: 62 SNP-based Haplotypes Associated with AMD Protection: 1,073 Cohort (927 Subset)

[00333] In addition to risk, several major CFH-to-F13B haplotypes showed protection from AMD relative to the cohort as a whole. H3_62_A (includes the rs800292 A allele) was significantly protective (Figure 6). Just as H1_62_A and H2_62_A differed in their risk association, so to did H3_62_A (protection) with the other major haplotype containing the rs800292 A Allele, H6_62_A (neutral). Of the haplotypes containing CFHR3/CFHR1 deletions, H5_62_A and H11_62_A showed significant protection, while H13_62_A showed marginal protection and H15_62_A was neutral. H3_62_A and H6_62_A are identical until mrd_3905/rs75816959, which is in CFHR5. Similarly the CFHR3/CFHR1 deletion containing haplotypes H5_62_A, H13_62_A, and H15_62_A are identical until mrd_3905. Thus, placing these haplotypes in the context of extended haplotypes that included CFHR5 and F13B resulted in significantly refined association with AMD (Figures 6 & 18).

[00334] 3.12 Results: 62 SNP-based Haplotypes Associated with Neutrality: GSH Cohort

[00335] Several major CFH-to-F13B haplotypes, including H4_62_A, H7_62_A, H10_62_A, H12_62_A, and H14_62_A, were neutral with respect to the development of AMD relative to the cohort as a whole. The minor allele of rs3753396 tagged many of these neutral haplotypes (Figure 1, Figure 3, Figure 6, and Figure 18). Other SNPs in LD with rs3753396 at a r^2 value of 90 or greater and that can be employed as surrogates for rs3753396 include (listed proximal to distal): rs10489456, rs70620, rs742855, rs11799380, rs1065489, rs11582939, rs385390, rs421820, rs426736, rs370953 and rs371075.

[00336] 3.13 Results: Gender Associations in 62 SNP-based Haplotypes: 1,073 Cohort (927 Subset)

[00337] 62 Single marker (SNPs) and haplotypes were examined for potential association with gender in the GSH case-control cohort. Associations were assessed using the entire cohort, the male sub-cohort only, and the female sub-cohort only (Figure 13 and Figure 23). The female cohort showed very similar levels of significance in the association of the 62 individual SNPs to AMD when compared to the '927 GSH Cohort (Figure 13). Male associations differed from the female associations. For example, association of rs800292, rs1061170 and rs1329421 was not observed in the male sub-cohort, a difference of approximately 11-orders of magnitude relative to females. Males did show lower levels of significant association with rs1410996, rs3753395 and rs393955, although also at levels up to 11-orders of magnitude less than was seen in the female cohort (Figure 13). The rs800292 A allele, previously associated with protection or reduced risk for AMD, also showed decreased association in the male sub-cohort. This data indicated that by controlling for sex, previous studies may have missed this stratification because of masking.

[00338] While the loss of association with AMD in males was significant throughout the CFH gene, one block of significant association remained, albeit at reduced levels. rs1410996 showed a p-value of 5.94×10^{-5} (compared to 3.26×10^{-16} in the full cohort and 6.31×10^{-13} in females) (Figure 13). This SNP was found to be in close to 100% LD with several proxy SNPs (rs2274700, rs3753395, and rs10737680). When looking at the female sub-cohort, rs1410996 had a similar strength of association to AMD as does rs1061170. It was the association of rs1410996 in males, an association lacking

at rs1061170, which accounted for the higher significance of rs1410996 (and its proxy SNPs rs2274700, rs3753395, and rs10737680). By analyzing males and females independently, a basis was identified for the disparity between risk at rs1061170 and rs1410996.

5 [00339] Studies suggested the male/female stratification seen at CFH was not a result of AMD disease in general, but of a specific mechanism of disease associated with CFH. Analysis of SNPs in C3 and C2/CFB showed much the same trend as with CFH: weaker but significant risk was found in both the full 927 cohort and the female-only cohort, while no risk was found in the male-only cohort. Thus, risk for
10 AMD in males was much less associated with the complement system as a whole than in females.

[00340] Haplotype disease associations were also examined independently in males and females. H2_62_A was barely significant in males but strongly significant in females (Figure 23). H2_51_B was more significant in males than H2_62_A, but still
15 less significant than in females (Figure 24). H1_62_A was significant in females, not at all in males. Whereas, H1_51_B was significant in both males and females; however, it was again more significant in females (Figure 24). H11_62_A was significant in both males and females, but was more significant in males. H2_62_A, H3_62_A, H5_62_A, and H11_62_A were all significantly associated with AMD in
20 all of the subjects (Figure 6) and also in the females (Figure 23). Females were slightly more significantly associated than the all-subjects in H1_62_A and H6_62_A, and less significantly associated in H13_62_A, but these differences were fairly minor.

[00341] 3.14 Results: 51 SNP-based Haplotypes Associated with AMD: 1,073
25 Cohort (927 Subset)

[00342] The association of haplotypes in the GSH 927 Cohort was also assessed using a 51 SNP-based haplotype analysis (Figure 22). The 51 SNPs included are depicted in Figure 22. The association of risk and protective haplotypes was similar to those observed in the 62 SNP-based haplotype analysis (Figure 6 and Figure 18;
30 also see Figure 20 and Figure 22).

[00343] 3.15 Results: 51 SNP-based Haplotypes and Their Association with AMD and AMD Phenotypes: ‘Combined Cohort’ and Extremely Discordant Sib-pair Cohort

[00344] A large case-control cohort comprised of individuals with and without AMD from the Iowa, Utah, and Melbourne cohorts (the ‘Combined Cohort’) was employed to assess 51 SNP-based CFH-to-F13B haplotypes with AMD (controls versus all AMD) risk, protection, and neutrality. Fifty-six haplotypes were generated from the Combined Cohort, 18 of them occurred at frequencies above 1% (Figure 15). The relationships between the major 51 SNP-based and 62 SNP-based haplotypes are shown in Figure 16 and the relationship between 51 SNP-based haplotypes in the 1,073 and Combined Cohorts is depicted in Figure 19. Associations similar to those of 62 SNP-based (Hx_62_A) and 51 SNP-based assessments in the GSH 927 Cohort (Hx_51_A) were observed (Figure 15). H1_51_B, however, became more significant in this larger cohort.

[00345] Several minor CFH-to-F13B haplotypes showed risk and protection from AMD relative to the cohort as a whole. For example, H24_62_A and H28_62_A trended toward risk and H30_62_A ($p=0.007$) and H40_62_A trended toward protection (Figure 15).

[00346] The ‘Combined Cohort’ was also employed to assess the association of single SNP associations of the 51 SNP panel and 51 SNP-based CFH-to-F13B haplotypes with the various phenotypes/stages of AMD, including comparisons of controls (stage 0) to all AMD (stages 1B-4C), early stage AMD (stages 1B-3), and the late stages of geographic atrophy (stage 4A) and CNV (stage 4B). Single marker associations in this cohort (Figure 42) did not differ from those of the 1,073 and highly discordant sib-pair cohorts when controls were compared to all AMD. Moreover, single marker associations with risk, protection and neutrality did not vary significantly when controls were compared to all AMD stages (Figure 42), early stage only (Figure 43), geographic atrophy only (Figure 44), or CNV only (Figure 45).

[00347] Haplotype associations with risk, protection and neutrality did not vary significantly between AMD phenotypes in the Combined Cohort (nor were they significantly different from analyses of the 1,073 cohort; Figure 20 and Figure 22) when controls were compared to early stage only, to geographic atrophy only, or to

CNV only (Figure 46). Note that the haplotype order (based on frequencies) was not identical in the three analyses shown in Figure 46..

[00348] The differential association of H1_51_B and H2_51_B with AMD risk was verified using a highly discordant sib-pair cohort. 51 SNP-based haplotypes were generated for this cohort. Using logistic regression, a pattern was seen in which H1_51_A (as described in the GSH cohort) was not significantly associated with risk. The equivalent to H2_51_A, namely H2_51_B, was highly associated with risk and was the only haplotype that associates with risk (Figure 12). Of the remaining haplotypes, only H5_51_B (containing the CFHR3/CFHR1 deletion) associated with AMD: in this case as a protective haplotype (Figure 12). Thus, the difference in risk association between H1_51_A and H2_51_A was been replicated in a second independent cohort.

[00349] 3.16 Results 51 SNP-based Diplotype Associations with AMD Risk, Protection and Neutrality: Combined Cohort

[00350] 51 SNP-based diplotypes were phased and determined in the Combined Cohort using BEAGLE version 3.3.2 (S R Browning and B L Browning, 2007, Rapid and accurate haplotype phasing and missing data inference for whole genome association studies using localized haplotype clustering. Am J Hum Genet 81:1084-1097). Association with AMD risk, protection, and neutrality was assessed in the entire cohort (Figure 35), in males only (Figure 36) and in females only (Figure 37). Combinations of all diplotype combinations for which there was appropriate statistical power are shown in Figure 35; statistical power was sufficient to assess associations through haplotype 7.

[00351] A single H1 haplotype when combined with another H1 haplotype, or a H2, H8 or H13 haplotype was associated with increased risk for developing AMD whereas combination of H1 haplotype with a H3 or H5 haplotype was associated with protection. Only when a single H2 haplotype occurred with another H2 haplotype, a H1 haplotype or a H10 haplotype was there an association with risk. No combinations showing association with protection were observed, suggesting that the H2 haplotype is more strongly associated with risk than is H1. No risk association was observed with any combinations of a single H3, H4, H5, H6 or H7 haplotype in

combination with any other haplotype. However, many of these combinations were protective, as depicted in Figure 35. The overall results for the male and female only groups were similar to those of the whole cohort, with some minor differences noted in males only, perhaps due to loss of statistical power (Figures 36 and 37).

5 **[00352]** The ‘Combined Cohort’ was also employed to assess the association of 51 SNP-based CFH-to-F13B diplotypes with the various phenotypes/stages of AMD, including comparisons of controls (stage 0) to all AMD (stages 1B-4C), to early stage AMD (stages 1B-3), and to CNV only (stage 4B). Diploidy associations with risk, protection and neutrality did not vary significantly between the following groups: 1) controls compared to all AMD stages (Figure 35); and 2) controls compared to CNV only (Figure 48). Nor did these associations differ from those in other cohorts. However, early when controls were compared to early stage AMD only (Figure 47), the protective effect of all H3 combinations was greatly diminished, suggesting that H3 diploidy combinations may not be protective for phenotypes (drusen) associated with early AMD development or with specific types of drusen associated with early AMD development.

[00353] 3.17 Results: Haplotype Tagging SNPs

20 **[00354]** Each of the major 62 SNP-based haplotypes and each of the major 51 SNP-based haplotypes can be uniquely identified using a combination of eight haplotype tagging SNPs (htSNPs; Figure 7; note that rs800292 and rs35928059 are optional for tagging the 62 and 51 SNP-based haplotypes). Case/control analysis was repeated on the GSH 1,073 Cohort using only the eight htSNPs to define haplotypes using Haploview 4.2. Risk association with H1 differed by several orders of magnitude and was statistically significant (Figure 10; 8 htSNPs – H1 p-value = 0.0024; 62 SNPs – H1 p-value = 0.116; note that 62 SNP-based significance values are slightly different in Figure 10 as compared to Figure 6 because Haploview cannot handle the tri-allelic deletion SNPs) compared to the 62 SNP-based analysis. In order to show this effect more clearly, the 927 GSH cohort was reanalyzed in Haploview using progressively fewer SNPs in the following numbers: 1) all CFH locus SNPs (62); 2) all SNPs minus the four deletion-associated SNPs (58); 3) all SNPs for which Taqman assays were developed (51); 4) all SNPs for which MAF >5% frequency (43); 6) all SNPs with

highly significant associations to AMD plus rs698859 (31); and 7) only the htSNPs (8) (Figure 10). As fewer SNPs were used to generate haplotypes, more minor haplotypes were recast as major haplotypes and H1 became significantly associated with risk for AMD. All other significant haplotypes showed only small changes in their association to AMD that did not affect the interpretation of risk.

[00355] 3.18 Results: Extension of 51 SNP-based CFH-to-F13B Haplotypes to Genome Level Haplotypes; Association of HapMap Haplotypes with AMD

[00356] 51 SNP-based haplotypes were compared to haplotypes generated using HapMap data in order to further refine haplotype association with AMD. The International HapMap Project database (<http://hapmap.ncbi.nlm.nih.gov/>) contains over 500 SNPs in the region encompassing the CFH-to-F13B locus (Hg18 chr1: 194869137-195303491). Genotypes from 180 Caucasian individuals (referred to as the CEU population) were downloaded for each of these 500 plus SNPs. Phased genotype data from the CEU set was downloaded, including the most informative SNPs (*i.e.* those that segregate in the CEU population) in this region of interest. Using Haploview (www.broadinstitute.org), haplotypes were constructed for the CEU dataset and their frequencies calculated (Figures 39 and 40). There is substantial overlap between the AMD case/control ('combined cohort') 51 SNP-based haplotypes and the HapMap haplotypes (Figures 38, 39 and 40). These overlapping SNPs were used as 'tagging' SNPs to match case/control haplotypes to their equivalent HapMap CEU haplotypes (Figures 38 and 41). AMD association with haplotype was previously established in the 51 SNP-based case/control ('combined') cohort (Figure 22); thus association could be extended to the equivalent HapMap haplotypes. In some instances, 51 SNP-based haplotypes overlapped with multiple HapMap haplotypes and vice versa due to the presence of HapMap SNPs that were not included in the 51 SNP-based haplotypes. Thus, in cases where HapMap haplotypes overlapped with multiple equivalent case/control haplotypes, key SNPs were identified that further refined the discriminative value of our major 51 SNP-based haplotypes. As an example, the 'combined cohort' haplotype H1 (Figures 39 and 41) matched the two most frequent of the major HapMap generated haplotypes (Figures 39 and 41). The only SNP that differentiated these two HapMap haplotypes was rs10494745. Other similar informative SNPs discovered in this analysis that will

refine assessment of disease association are indicated with asterisks and their relationship to the Combined Cohort 51 SNP-based haplotypes are described in Figure 40. They are rs7524776, rs7513157, rs419137, and rs10494745.

[00357] 3.19 Results: Extended Haplotype Blocks

[00358] It was determined whether there were SNPs proximal and distal to the boundaries defined by the 62 SNP-based haplotypes that are in LD with the defined major haplotype blocks. All Hapmap SNPs for Caucasians lying between Chr1:194,814,311 and 195,431,890 were downloaded with subject information and analyzed in Haploview (Figures 14). Default (Gabriel et al.) blocks encompassed the entire region. The second block, defined by rs10801551 (194,848,662) through rs800292 (194,908,856) encompassed rs35928059, the most 5' SNP employed in the 62-SNP set. This suggests that the rs35928059 is in LD with SNPs as far proximal as rs10801551. Similarly, at the distal 3' end of our locus, rs6428380, the most 3' SNP in the 62_A SNP set, lies between rs4915148 (195,302,161) and rs1537319 (195,319,427), suggesting that the distal SNPs within this block are likely in LD with rs1537319.

[00359] 3.20 Results: Characterization of CFHR3/CFHR1 Deletion Boundaries

[00360] Quantitative PCR (qPCR) was performed on multiple CEPH individuals representing each deletion tagging SNP genotype. Three qPCR primer sets were designed to amplify both duplicated regions equally, and two additional probes were designed in the sequence between the segmental duplications. All experiments were performed using QuantiFast SYBR RT-PCR Master Mix (Qiagen) and the MyiQ Single Color Real-Time PCR Detection System (BioRad). Non-deletion homozygotes showed the presence of both segmental duplications, as well as normal amplification of probes between the duplications. Heterozygotes showed intermediate values for probes within the duplications and a one-copy deletion in the region between the duplications. Individuals homozygous for the deletion tagging allele showed the presence of only one segmental duplication region and no amplification of probes located between the duplicated regions. Thus this qPCR assay can be used to reliably identify individuals as homozygous R3/R1 deletion, heterozygous R3/R1 deletion, or homozygous R3/R1 non-deletion.

[00361] 3.21 Results: Development of Alternative CFHR3/CFHR1 Deletion Status Assays and Confirmation of rs12144939 as a CFHR3/CFHR1 Deletion Tagging SNP

[00362] Two additional PCR assays were developed to confirm presence of deletion or presence on non-deletion chromosomes (Figure 11). These assays were not designed to distinguish heterozygotes, but to determine status of homozygous deletion or non-deletion samples. Using the sequence variant signatures found in deletion homozygotes, primers were designed to generate a 3.7kb PCR product only when the deletion is present, either in heterozygous or homozygous form. Reactions were performed using AccuPrime High Fidelity polymerase, buffers, and protocols (Invitrogen). Samples from twelve unrelated CEPH individuals and five African individuals were amplified and run on a 1% agarose gel. All homozygous deletion and heterozygous individuals produced a 3.7kb band in the assay designed to amplify across the deletion, while none of the homozygote non-deletion individuals amplified. In addition, primers were designed and used to amplify a small (200bp) PCR product only when the deletion is not present. Reactions were performed on eighty-eight CEPH individuals. All amplified well except for deletion homozygotes, which failed without exception. Thus these two assays can be used to unambiguously identify individuals homozygous either for the CFHR3-R1 deletion or for non-deletion status and in combination can determine heterozygotes.

[00363] 3.21 Confirmation of the Deletion Tagging SNP

[00364] A predesigned Taqman CNV assay was run on all 604 CEPH individuals in order to confirm deletion status. This assay was located on chr1:195011298-195011397, just proximal to the *CFHR3* gene. Assays were run on a 7900HT Fast Real-Time PCR System (Applied Biosystems) and analyzed using CopyCaller software (Applied Biosystems). Copy number calls were compared against rs12144939 (deletion tagging SNP) genotypes. In all cases where a clear call was made by CopyCaller software, copy number calls were consistent with what would be predicted by rs12144939, with the exception of one family in which six individuals were heterozygous for the tagging SNP but were called homozygous non-deletion in the CNV analysis. This confirmed that in Caucasians, rs12144939 tagged the deletion with near 100% reliability. Thus, CFHR1/R3 deletion status can now be obtained

using the three assays described above. Analysis of the 1000-genome data indicates that the following SNPs accurately tag rs12144939, and therefore the CFHR3/CFHR1 deletion: rs6689009, rs35253683, rs731557, and rs60642321.

5 [00365] It will be apparent to those skilled in the art that various modifications and variations can be made in the present invention without departing from the scope or spirit of the invention. Other aspects of the invention will be apparent to those skilled in the art from consideration of the specification and practice of the invention described herein. It is intended that the specification and examples be considered as exemplary only, with a true scope and spirit of the invention being indicated by the
10 following claims.

[00366] Throughout this application, various publications are referenced. The disclosures of these publications in their entireties are hereby incorporated by reference into this application in order to more fully describe the state of the art to which this pertains. The references disclosed are also individually and specifically
15 incorporated by reference herein for the material contained in them that is discussed in the sentence in which the reference is relied upon. Nothing herein is to be construed as an admission that the present invention is not entitled to antedate such publication by virtue of prior invention. Further, the dates of publication provided herein may be different from the actual publication dates, which can require
20 independent confirmation.

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CLAIMS

What is claimed is:

1. A method for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates the subject's susceptibility for having or developing age-related macular degeneration.
2. The method of claim 1, wherein the subject's haplotype is determined from a sample obtained from the subject.
3. The method of claim 1, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.
4. The method of claim 1, wherein H2_62_A, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration.
5. The method of claim 4, further comprising administering a therapeutic composition to the subject.
6. The method of claim 1, wherein H3_62_A, H5_62_A, H11_62_A, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration.
7. The method of claim 1, wherein the subject is female.
8. The method of claim 7, wherein H1_62_A, H2_62_A, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration.
9. The method of claim 7, wherein H3_62_A, H5_62_A, H11_62_A, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

10. The method of claim 1, wherein the subject is male.

11. The method of claim 10, wherein H2_62_A, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration

12. The method of claim 10, wherein H11_62_A, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

13. A method for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates the subject's susceptibility for having or developing age-related macular degeneration.

14. The method of claim 13, wherein the subject's haplotype is determined from a sample obtained from the subject.

15. The method of claim 13, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

16. The method of claim 13, wherein H2_51_A, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration.

17. The method of claim 16, further comprising administering a therapeutic composition to the subject.

18. The method of claim 13, wherein H3_51_A, H5_51_A, H10_51_A, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

19. A method for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B,

H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates the subject's susceptibility for having or developing age-related macular degeneration.

20. The method of claim 19, wherein the subject's haplotype is determined from a sample obtained from the subject.

21. The method of claim 19, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

22. The method of claim 19, wherein H1_51_B, H2_51_B, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration.

23. The method of claim 22, further comprising administering a therapeutic composition to the subject.

24. The method of claim 16, wherein H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

25. The method of claim 19, wherein the subject is female.

26. The method of claim 25, wherein H1_51_B, H2_51_B, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration

27. The method of claim 25, wherein H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration

28. The method of claim 19, wherein the subject is male.

29. The method of claim 28, wherein H1_51_B, H2_51_B, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration.

30. The method of claim 28, wherein H3_51_B, H14_51_B, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

31. A method for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus, wherein the SNPs are: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of at least six of the SNPs indicates the subject's susceptibility for having or developing age-related macular degeneration.

32. The method of claim 31, wherein the subject's haplotype is determined from a sample obtained from the subject.

33. The method of claim 31, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

34. The method of claim 31, wherein the at least six SNPs consists of at least rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859.

35. The method of claim 31, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

36. The method of claim 31, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

37. The method of claim 31, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

38. The method of any of claims 35-37, further comprising administering a therapeutic composition to the subject.

39. The method of claim 31, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

40. The method of claim 31, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

41. The method of claim 31, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

42. The method of claim 31, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

43. The method of claim 31, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

44. The method of claim 31, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

45. The method of claim 31, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T

at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

46. The method of claim 31, wherein the subject is female.

47. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an G at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

48. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

49. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

50. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

51. The method of claim 46, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

52. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

53. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

54. The method of claim 46, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

55. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

56. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

57. The method of claim 46, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

58. The method of claim 31, wherein the subject is male.

59. The method of claim 58, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

60. The method of claim 58, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

61. The method of claim 58, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 is indicative of the subject's increased risk for having or developing age-related macular degeneration.

62. The method of claim 58, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

63. The method of claim 58, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

64. The method of claim 58, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

65. A method for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus, wherein the SNPs is: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of one or more of the SNPs indicates the subject's susceptibility for having or developing age-related macular degeneration.

66. The method of claim 65, wherein the subject's haplotype is determined from a sample obtained from the subject.

67. The method of claim 65, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

68. The method of claim 65, wherein an A at the rs1061170 SNP is indicative of the subject's increased risk for having or developing age-related macular degeneration.

69. The method of claim 65, wherein an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

70. A method for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus, wherein the SNP is: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939, and wherein the presence of the SNP indicates the subject's susceptibility for having or developing age-related macular degeneration.

71. The method of claim 70, wherein the subject's haplotype is determined from a sample obtained from the subject.

72. The method of claim 70, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

73. The method of claim 70, wherein a T at the rs12144939 SNP is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

74. A method of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of treatment for age-related macular degeneration.

75. The method of claim 74, wherein the subject's haplotype is determined from a sample obtained from the subject.

76. The method of claim 74, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

77. The method of claim 74, wherein H2_62_A, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

78. The method of claim 74, wherein H3_62_A, H5_62_A, H11_62_A, or a complement thereof, indicates the subject is not in need of treatment for age-related macular degeneration.

79. The method of claim 74, wherein H1_62_A, H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, indicates the subject may be in need of treatment for age-related macular degeneration.

80. The method of claim 74, wherein the subject is a female.

81. The method of claim 80, wherein H1_62_A, H2_62_A, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

82. The method of claim 80, wherein H3_62_A, H5_62_A, H11_62_A, or a complement thereof, indicates the subject is not in need of treatment for age-related macular degeneration.

83. The method of claim 80, wherein H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, indicates the subject may be in need of treatment for age-related macular degeneration.

84. The method of claim 74, wherein the subject is male.

85. The method of claim 84, wherein H2_62_A, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

86. The method of claim 84, wherein H11_62_A, or a complement thereof, indicates the subject is not in need of treatment for age-related macular degeneration.

87. The method of claim 84, wherein H1_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, indicates the subject may be in need of treatment for age-related macular degeneration.

88. A method of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of treatment for age-related macular degeneration.

89. The method of claim 88, wherein the subject's haplotype is determined from a sample obtained from the subject.

90. The method of claim 88, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

91. The method of claim 88, wherein H2_51_A, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

92. The method of claim 88, wherein H3_51_A, H5_51_A, H10_51_A, or a complement thereof, indicates the subject is not in need of treatment for age-related macular degeneration.

93. The method of claim 88, wherein H1_51_A, H4_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, indicates the subject may be in need of treatment for age-related macular degeneration.

94. A method of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of treatment for age-related macular degeneration.

95. The method of claim 94, wherein the subject's haplotype is determined from a sample obtained from the subject.

96. The method of claim 94, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

97. The method of claim 94, wherein H1_51_B, H2_51_B, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

98. The method of claim 94, wherein H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof, indicates the subject is not in need of treatment for age-related macular degeneration.

99. The method of claim 94, wherein H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, indicates the subject may be in need of treatment for age-related macular degeneration.

100. The method of claim 94, wherein the subject is female.

101. The method of claim 100, wherein H1_51_B, H2_51_B, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

102. The method of claim 100, wherein H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof, indicates the subject is not in need of treatment for age-related macular degeneration.

103. The method of claim 100, wherein H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, or a complement thereof, indicates the subject may be in need of treatment for age-related macular degeneration.

104. The method of claim 94, wherein the subject is male.

105. The method of claim 104, wherein H1_51_B, H2_51_B, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

106. The method of claim 104, wherein H3_51_B, H14_51_B, or a complement thereof, indicates the subject is not in need of treatment for age-related macular degeneration.

107. The method of claim 104, wherein H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, indicates the subject may be in need of treatment for age-related macular degeneration.

108. A method of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus, wherein the SNPs are: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of at least six of the SNPs indicates whether the subject is in need of treatment for age-related macular degeneration.

109. The method of claim 108, wherein the subject's haplotype is determined from a sample obtained from the subject.

110. The method of claim 108, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

111. The method of claim 108, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 indicates the subject is in need of treatment for age-related macular degeneration.

112. The method of claim 108, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 indicates the subject is in need of treatment for age-related macular degeneration.

113. The method of claim 108, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 indicates the subject is in need of treatment for age-related macular degeneration.

114. The method of claim 108, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 indicates the subject is not in need of treatment for age-related macular degeneration.

115. The method of claim 108, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 indicates the subject is not in need of treatment for age-related macular degeneration.

116. The method of claim 108, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 indicates the subject is not in need of treatment for age-related macular degeneration.

117. The method of claim 108, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 indicates the subject is not in need of treatment for age-related macular degeneration.

118. The method of claim 108, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 indicates the subject is not in need of treatment for age-related macular degeneration.

119. The method of claim 108, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 indicates the subject is not in need of treatment for age-related macular degeneration.

120. The method of claim 108, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 indicates the subject is not in need of treatment for age-related macular degeneration.

121. A method of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus, wherein the SNPs is: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of one or more of the SNPs indicates whether the subject is in need of treatment for age-related macular degeneration.

122. The method of claim 121, wherein the subject's haplotype is determined from a sample obtained from the subject.

123. The method of claim 121, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

124. The method of claim 121, wherein an A at the rs1061170 SNP indicates the subject is in need of treatment for age-related macular degeneration.

125. The method of claim 121, wherein an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP indicates the subject is not in need of treatment for age-related macular degeneration.

126. The method of claim 121, wherein a G at the rs3753396 SNP indicates the subject may be in need of treatment for age-related macular degeneration.

127. A method of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus, wherein the SNPs are: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939, and wherein the presence of one or more of the SNPs indicates whether the subject is in need of treatment for age-related macular degeneration.

128. The method of claim 127, wherein the subject's haplotype is determined from a sample obtained from the subject.

129. The method of claim 127, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

130. The method of claim 127, wherein a T at the rs12144939 SNP indicates the subject is not in need of treatment for age-related macular degeneration.

131. A method of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

132. The method of claim 131, wherein the subject's haplotype is determined from a sample obtained from the subject.

133. The method of claim 131, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

134. The method of claim 131, wherein H2_62_A, or a complement thereof, indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

135. The method of claim 131, wherein H3_62_A, H5_62_A, H11_62_A, or a complement thereof, indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

136. The method of claim 131, wherein H1_62_A, H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, indicates the subject may be in need of prophylactic treatment for age-related macular degeneration.

137. A method of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the

haplotypes indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

138. The method of claim 137, wherein the subject's haplotype is determined from a sample obtained from the subject.

139. The method of claim 137, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

140. The method of claim 137, wherein H2_51_A, or a complement thereof, indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

141. The method of claim 137, wherein H3_51_A, H5_51_A, H10_51_A, or a complement thereof, indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

142. The method of claim 137, wherein H1_51_A, H4_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, indicates the subject may be in need of prophylactic treatment for age-related macular degeneration.

143. A method of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

144. The method of claim 143, wherein the subject's haplotype is determined from a sample obtained from the subject.

145. The method of claim 143, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

146. The method of claim 143, wherein H1_51_B, H2_51_B, or a complement thereof, indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

147. The method of claim 143, wherein H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof, indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

148. The method of claim 143, wherein H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, indicates the subject may be in need of prophylactic treatment for age-related macular degeneration.

149. A method of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus, wherein the SNPs are: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of at least six of the SNPs indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

150. The method of claim 149, wherein the subject's haplotype is determined from a sample obtained from the subject.

151. The method of claim 149, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

152. The method of claim 149, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

153. The method of claim 149, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T

at rs698859 indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

154. The method of claim 149, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

155. The method of claim 149, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

156. The method of claim 149, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

157. The method of claim 149, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

158. The method of claim 149, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

159. The method of claim 149, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

160. The method of claim 149, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a

T at rs698859 indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

161. The method of claim 149, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

162. A method of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus, wherein the SNPs is: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of one or more of the SNPs indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

163. The method of claim 162, wherein the subject's haplotype is determined from a sample obtained from the subject.

164. The method of claim 162, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

165. The method of claim 162, wherein an A at the rs1061170 SNP indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

166. The method of claim 162, wherein an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

167. The method of claim 162, wherein a G at the rs3753396 SNP indicates the subject may be in need of prophylactic treatment for age-related macular degeneration.

168. A method of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus, wherein the SNPs are: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939, and wherein the presence of one or more

of the SNPs indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

169. The method of claim 168, wherein the subject's haplotype is determined from a sample obtained from the subject.

170. The method of claim 168, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

171. The method of claim 168, wherein a T at the rs12144939 SNP indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

172. A method of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is appropriate for the clinical trial.

173. The method of claim 172, wherein the subject's haplotype is determined from a sample obtained from the subject.

174. The method of claim 172, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

175. The method of claim 172, wherein H2_62_A, or a complement thereof, indicates the subject is appropriate for the clinical trial.

176. The method of claim 172, wherein H3_62_A, H5_62_A, H11_62_A, or a complement thereof, indicates the subject is not appropriate for the clinical trial.

177. The method of claim 172, wherein H1_62_A, H4_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H12_62_A, H13_62_A, H14_62_A, H15_62_A, or a complement thereof, indicates the subject may be appropriate for the clinical trial.

178. A method of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is appropriate for the clinical trial.

179. The method of claim 178, wherein the subject's haplotype is determined from a sample obtained from the subject.

180. The method of claim 178, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

181. The method of claim 178, wherein H2_51_A, or a complement thereof, indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

182. The method of claim 178, wherein H3_51_A, H5_51_A, H10_51_A, or a complement thereof, indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

183. The method of claim 178, wherein H1_51_A, H4_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, or a complement thereof, indicates the subject may be in need of prophylactic treatment for age-related macular degeneration.

184. A method of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the Caucasian subject the identity of one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the haplotypes indicates whether the subject is appropriate for the clinical trial.

185. The method of claim 184, wherein the subject's haplotype is determined from a sample obtained from the subject.

186. The method of claim 184, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

187. The method of claim 184, wherein H1_51_B, H2_51_B, or a complement thereof, indicates the subject is appropriate for the clinical trial.

188. The method of claim 184, wherein H3_51_B, H5_51_B, H12_51_B, H14_51_B, or a complement thereof, indicates the subject is not appropriate for the clinical trial.

189. The method of claim 184, wherein H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, indicates the subject may be appropriate for the clinical trial.

190. A method of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the Caucasian subject the identity of at least six SNPs in the CFH-to-F13B locus, wherein the SNPs are: (i) rs35928059, rs800292, rs1061170, rs12144939, rs7546940, rs1409153, rs10922153, or rs698859, or (ii) a SNP in linkage disequilibrium with the SNPs of (i), and wherein the presence of at least six of the SNPs indicates whether the subject is appropriate for the clinical trial.

191. The method of claim 190, wherein the subject's haplotype is determined from a sample obtained from the subject.

192. The method of claim 190, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

193. The method of claim 190, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a G at rs1409153, a G at rs10922153, or an A at rs698859 indicates the subject is appropriate for the clinical trial.

194. The method of claim 190, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a T at rs698859 indicates the subject is appropriate for the clinical trial.

195. The method of claim 190, wherein an A at rs35928059, a G at rs800292, a C at rs1061170, a G at rs12144939, a G at rs7546940, a C at rs1409153, a G at rs10922153, or a C at rs698859 indicates the subject is appropriate for the clinical trial.

196. The method of claim 190, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 indicates the subject is not appropriate for the clinical trial.

197. The method of claim 190, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, an A at rs1409153, a T at rs10922153, or a G at rs698859 indicates the subject is not appropriate for the clinical trial.

198. The method of claim 190, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, an A at rs1409153, a T at rs10922153, or an A at rs698859 indicates the subject is not appropriate for the clinical trial.

199. The method of claim 190, wherein an A at rs35928059, an A at rs800292, a T at rs1061170, a G at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 indicates the subject is not appropriate for the clinical trial.

200. The method of claim 190, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a C at rs698859 indicates the subject is not appropriate for the clinical trial.

201. The method of claim 190, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, an A at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 indicates the subject is not appropriate for the clinical trial.

202. The method of claim 190, wherein an A at rs35928059, a G at rs800292, a T at rs1061170, a T at rs12144939, a G at rs7546940, a T at rs1409153, a T at rs10922153, or a T at rs698859 indicates the subject is not appropriate for the clinical trial.

203. A method of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the subject the identity of one or more SNPs in the CFH-to-F13B locus, wherein the SNPs is: (i) rs1061170, rs1410996, rs2274700, rs3753395, rs403846, or rs3753396 or (ii) a SNP in linkage disequilibrium with the SNPs of

(i), and wherein the presence of one or more of the SNPs indicates whether the subject is appropriate for the clinical trial.

204. The method of claim 203, wherein the subject's haplotype is determined from a sample obtained from the subject.

205. The method of claim 203, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

206. The method of claim 203, wherein an A at the rs1061170 SNP indicates the subject is appropriate for the clinical trial.

207. The method of claim 203, wherein an A at the rs1410996 SNP, an A at the rs2274700 SNP, a T at the rs3753395 SNP, or a G at the rs403846 SNP indicates the subject is not appropriate for the clinical trial.

208. The method of claim 203, wherein a G at the rs3753396 SNP indicates the subject may be appropriate for the clinical trial.

209. A method of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the subject the identity of a deletion tagging SNP in the CFH-to-F13B locus, wherein the SNPs are: (i) rs12144939 or (ii) a SNP in linkage disequilibrium with rs12144939, and wherein the presence of one or more of the SNPs indicates whether the subject is appropriate for the clinical trial.

210. The method of claim 209, wherein the subject's haplotype is determined from a sample obtained from the subject.

211. The method of claim 209, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

212. The method of claim 209, wherein a T at the rs12144939 SNP indicates the subject is not appropriate for the clinical trial.

213. A kit comprising an assay for detecting one or more haplotypes in a nucleic acid sample of a subject, wherein the one or more haplotypes are H1_62_A, H2_62_A, H3_62_A, H4_62_A, H5_62_A, H6_62_A, H7_62_A, H8_62_A, H9_62_A, H10_62_A, H11_62_A,

H12_62_A, H13_62_A, H14_62_A, H15_62_A, H1_51_A, H2_51_A, H3_51_A, H4_51_A, H5_51_A, H6_51_A, H7_51_A, H8_51_A, H9_51_A, H10_51_A, H11_51_A, H12_51_A, H13_51_A, H14_51_A, H15_51_A, H16_51_A, H17_51_A, H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof.

214. The kit of claim 213, further comprising amplification reagents for amplifying the CHF-to-F13B locus.

215. The kit of claim 213, further comprising instructions for correlating the assay results with the subject's risk for having or developing age-related macular degeneration.

216. A kit comprising an assay for detecting one or more SNPs in a nucleic acid sample of a subject, wherein the SNP is (i) rs12144939, rs1061170, rs1410996, rs2274700, rs3753395, or rs403846; (ii) combinations of the SNPs of (i); or (iii) a SNP in linkage disequilibrium with the SNPs of (i).

217. The kit of claim 216, further comprising amplification reagents for amplifying the CHF-to-F13B locus.

218. The kit of claim 216, further comprising instructions for correlating the assay results with the subject's risk for having or developing age-related macular degeneration.

219. A method for determining a Caucasian subject's susceptibility to having or developing age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates the subject's susceptibility for having or developing age-related macular degeneration.

220. The method of claim 219, wherein the subject's haplotype is determined from a sample obtained from the subject.

221. The method of claim 219, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

222. The method of claim 219, wherein the diplotype H1_51_B: H1_51_B, H1_51_B:H2_51_B, H1_51_B:H8_51_B, H1_51_B:H13_51_B, H2_51_B:H2_51_B, H2_51_B:H10_51_B, H1_51_B:any minor 51_B haplotype, H2_51_B:any minor 51_B haplotype, or a complement thereof, is indicative of the subject's increased risk for having or developing age-related macular degeneration.

223. The method of claim 222, further comprising administering a therapeutic composition to the subject.

224. The method of claim 219, wherein diplotype H1_51_B: H3_51_B, H1_51_B:H5_51_B, H1_51_B:H14_51_B, H3_51_B: H3_51_B, H3_51_B: H4_51_B, H3_51_B: H5_51_B, H3_51_B: H6_51_B, H3_51_B: any 51_B minor haplotype, H4_51_B: H5_51_B, H5_51_B: H5_51_B, H5_51_B: any 51_B minor haplotype, H6_51_B: H6_51_B, H6_51_B: any 51_B minor haplotype, or a complement thereof, is indicative of the subject's decreased risk for having or developing age-related macular degeneration.

225. A method of identifying a Caucasian subject in need of treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates whether the subject is in need of treatment for age-related macular degeneration.

226. The method of claim 225, wherein the subject's haplotype is determined from a sample obtained from the subject.

227. The method of claim 225, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

228. The method of claim 225, wherein diplotype H1_51_B: H1_51_B, H1_51_B:H2_51_B, H1_51_B:H8_51_B, H1_51_B:H13_51_B, H2_51_B:H2_51_B,

H2_51_B:H10_51_B, H1_51_B: any minor 51_B haplotype, H2_51_B: any minor 51_B haplotype, or a complement thereof, indicates the subject is in need of treatment for age-related macular degeneration.

229. The method of claim 225, wherein diplotype H1_51_B: H3_51_B, H1_51_B:H5_51_B, H1_51_B:H14_51_B, H3_51_B: H3_51_B, H3_51_B: H4_51_B, H3_51_B: H5_51_B, H3_51_B: H6_51_B, H3_51_B: any 51_B minor haplotype, H4_51_B: H5_51_B, H5_51_B: H5_51_B, H5_51_B: any 51_B minor haplotype, H6_51_B: H6_51_B, H6_51_B: any 51_B minor haplotype, or a complement thereof, indicates the subject is not in need of treatment for age-related macular degeneration.

230. The method of claim 225, wherein diplotype H4_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H13_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, indicates the subject may be in need of treatment for age-related macular degeneration.

231. A method of identifying a Caucasian subject in need of a prophylactic treatment for age-related macular degeneration comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates whether the subject is in need of prophylactic treatment for age-related macular degeneration.

232. The method of claim 231, wherein the subject's haplotype is determined from a sample obtained from the subject.

233. The method of claim 231, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

234. The method of claim 231, wherein diplotype H1_51_B: H1_51_B, H1_51_B:H2_51_B, H1_51_B:H8_51_B, H1_51_B:H13_51_B, H2_51_B:H2_51_B, H2_51_B:H10_51_B, H1_51_B: any minor 51_B haplotype, H2_51_B: any minor 51_B

haplotype, or a complement thereof, indicates the subject is in need of prophylactic treatment for age-related macular degeneration.

235. The method of claim 231, wherein diplotype H1_51_B: H3_51_B, H1_51_B:H5_51_B, H1_51_B:H14_51_B, H3_51_B: H3_51_B, H3_51_B: H4_51_B, H3_51_B: H5_51_B, H3_51_B: H6_51_B, H3_51_B: any 51_B minor haplotype, H4_51_B: H5_51_B, H5_51_B: H5_51_B, H5_51_B: any 51_B minor haplotype, H6_51_B: H6_51_B, H6_51_B: any 51_B minor haplotype, or a complement thereof, indicates the subject is not in need of prophylactic treatment for age-related macular degeneration.

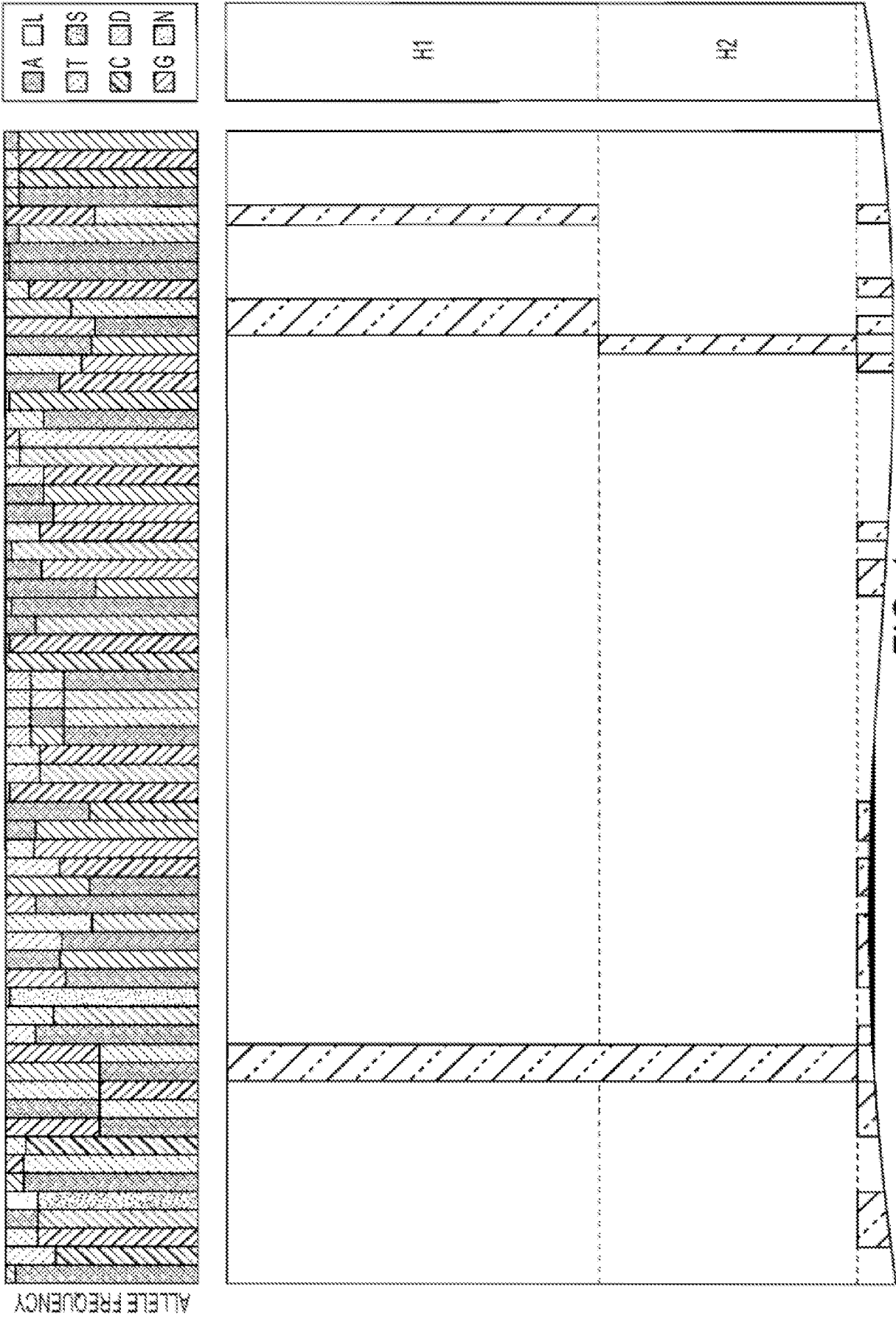
236. A method of identifying a Caucasian subject appropriate for an age-related macular degeneration clinical trial comprising determining in the Caucasian subject the identity of one or more diplotypes based on one or more haplotypes, wherein the one or more haplotypes are H1_51_B, H2_51_B, H3_51_B, H4_51_B, H5_51_B, H6_51_B, H7_51_B, H8_51_B, H9_51_B, H10_51_B, H11_51_B, H12_51_B, H13_51_B, H14_51_B, H15_51_B, H16_51_B, H17_51_B, H18_51_B, H19_51_B, H20_51_B, H21_51_B, H22_51_B, or a complement thereof, and wherein the presence of one or more of the diplotypes indicates whether the subject is appropriate for the clinical trial.

237. The method of claim 236, wherein the subject's haplotype is determined from a sample obtained from the subject.

238. The method of claim 236, wherein the subject's haplotype is determined by amplifying or sequencing a nucleic acid sample obtained from the subject.

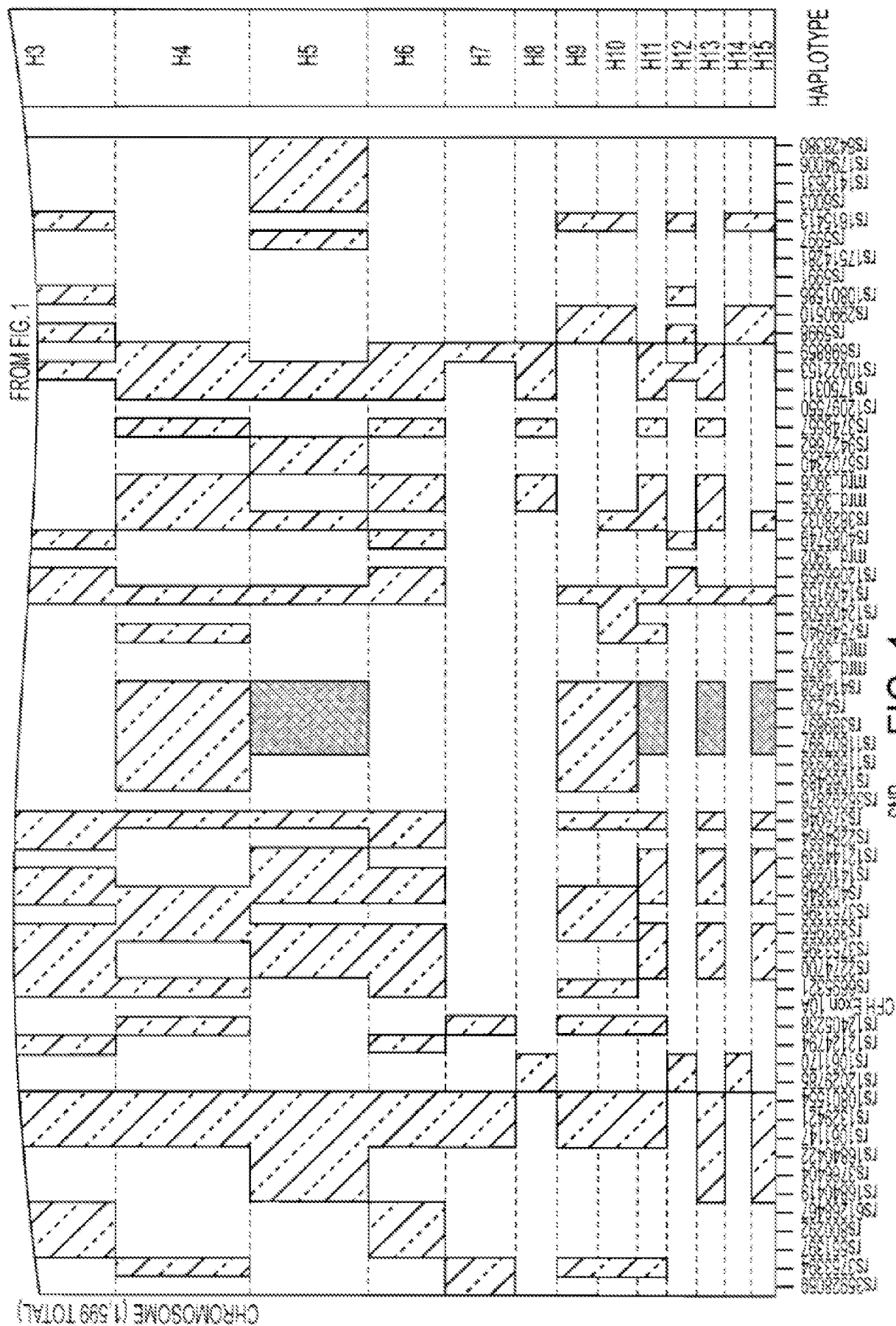
239. The method of claim 236, wherein diplotype H1_51_B: H1_51_B, H1_51_B:H2_51_B, H1_51_B:H8_51_B, H1_51_B:H13_51_B, H2_51_B:H2_51_B, H2_51_B:H10_51_B, H1_51_B: any minor 51_B haplotype, H2_51_B: any minor 51_B haplotype, or a complement thereof, indicates the subject is appropriate for the clinical trial.

240. The method of claim 236, wherein diplotype H1_51_B: H3_51_B, H1_51_B:H5_51_B, H1_51_B:H14_51_B, H3_51_B: H3_51_B, H3_51_B: H4_51_B, H3_51_B: H5_51_B, H3_51_B: H6_51_B, H3_51_B: any 51_B minor haplotype, H4_51_B: H5_51_B, H5_51_B: H5_51_B, H5_51_B: any 51_B minor haplotype, H6_51_B: H6_51_B, H6_51_B: any 51_B minor haplotype, or a complement thereof, indicates the subject is not appropriate for the clinical trial.



TO FIG. 1 CONTINUED

FIG. 1



	SNP	Gene	Context		Position GRCh37	Hetero- zygosity	GSH Minor Allele	MAF	alt. ID
1	rs35928059	5' of CFH		A/G	196620471	0.043	G	0.051	
2	rs3753394	5' of CFH		C/T	196620917	0.415	T	0.264	
3	rs551397	CFH	intron 1	C/T	196642072	0.485	T	0.165	
4	rs800292	CFH	V62I	A/G	196642233	0.495	A	0.165	
5	rs61268467	CFH	intron 2	-/T	196642978:196642979				rs55714531
6	rs16840419	CFH	intron 6	A/G	196651745	0.249	G	0.107	
7	rs3766404	CFH	intron 6	C/T	196651832	0.38	C	0.105	
8	rs16840422	CFH	intron 6	C/T	196652834	0.384	T	0.109	
9	rs1061147	CFH	A307A exon7	A/C	196654324	0.403	C	0.49	
10	rs1329421	CFH	intron 7	A/T	196656205	0.38	A	0.49	
11	rs10801554	CFH	intron 7	C/T	196657655	0.331	T	0.49	
12	rs12029785	CFH	intron 8	A/G	196658905	0.404	G	0.49	
13	rs1061170	CFH	H402Y exon9	C/T	196659237		C	0.49	
14	rs12124794	CFH	intron 9	A/T	196661538	0.312	T	0.152	
15	rs12405238	CFH	intron 9	G/T	196661613	0.401	T	0.249	
16	CFH_Exon_10A	CFH	intron 9	-/GG	196670506			0.005	
17	rs6695321	CFH	intron 9	A/G	196675861	0.497	G	0.311	
18	rs2274700	CFH	A473A exon10	A/G	196682947	0.496	A	0.303	
19	rs3753395	CFH	intron 11	A/T	196686652	0.499	T	0.306	
20	rs393955	CFH	intron 11	G/T	196692470	0.368	T	0.465	
21	rs3753396	CFH	Q672Q exon13	A/G	196695742	0.421	G	0.158	
22	rs403846	CFH	intron 14	A/G	196696737	0.336	G	0.465	
23	rs1410996	CFH	intron 14	C/T	196696933	0.498	T	0.307	
24	rs12144939	CFH	intron 15	A/G/T	196698945		T	0.154	
25	rs2284664	CFH	intron 15	A/G	196702525	0.427	A	0.151	
26	rs375046	CFH	intron 15	A/C	196705926	0.359	A	0.465	
27	rs35292876	CFH	H878H exon17	C/T	196706642	0.022	T	0.01	
28	rs1065489	CFH	E936D exon18	G/T	196709774	0.489	T	0.163	
29	rs11582939	CFH	intron 18	C/T	196710157	0.382	T	0.164	
30	rs11807997	5' of CFHR3	DEL	A/G	196743213	0.5	G	0.192	
31	rs389897	CFHR3	intron 4 DEL	A/T	196758430	0.482	A	0.194	
32	rs4230	CFHR1	R302R exon6 DEL	G/T	196801042	0.364	T	0.197	

FIG. 2

33	rs414628	CFHR1	R314R exon6 DEL	A/T	196801078	0.5	T	0.197	
34	mrd_3876	5' of CFHR4		C/G	196856942		G	0.012	rs112797613
35	mrd_3877	5' of CFHR4		C/A	196857150		A	0.01	
36	rs7546940	CFHR4	intron 1	A/G	196870792	0.321	A	0.154	
37	rs12406509	CFHR4	intron 2	A/G	196874192	0.02	G	0.026	mrd_3878
38	rs1409153	CFHR4	intron 2	A/G	196880005		A	0.49	
39	rs432366	5' of CFHR2		C/G	196912191		C		
40	rs12066959	CFHR2	intron 1	A/G	196917899		A	0.176	
41	mrd_3902	CFHR2	C72Y exon2	A/G	196918741	0.027	A	0.018	rs79351096
42	rs4085749	CFHR2	C140C exon3	C/T	196920148	0.448	T	0.176	
43	rs3828032	CFHR2	intron 3	A/G	196920178	0.424	A	0.258	
44	mrd_3905	5' of CFHR5		G/A	196944722		A	0.197	rs75816959
45	mrd_3906	5' of CFHR5		C/T	196944860		T	0.199	rs74188363
46	rs6702340	5' of CFHR5		G/T	196945050	0.289	T	0.08	
47	rs9427662	CFHR5	5'UTR	C/T	196946775	0.294	C	0.078	
48	rs3748557	CFHR5	intron 1	A/T	196946869	0.253	T	0.201	
49	rs12097550	CFHR5	P46S exon2	C/T	196952092	0.013	T	0.004	
50	rs1750311	CFHR5	intron 3	A/C	196954225		A	0.28	
51	rs10922153	CFHR5	3'UTR	G/T	196978615	0.409	T	0.409	
52	rs698859	F13B	3'UTR	A/G	197008365	0.351	A	0.446	
53	rs5998	F13B	N602N exon 11	A/G	197009798	0.497	G	0.474	
54	rs2990510	F13B	intron 9	G/T	197020658	0.302	G	0.349	
55	rs10801586	F13B	intron 9	C/T	197021189	0.388	T	0.125	
56	rs5991	F13B	E388V exon7	A/T	197026151		T	0.008	
57	rs17514281	F13B	I342T exon7	C/T	197026289	0.022	C	0.008	
58	rs5997	F13B	T152T exon4	A/G	197030201	0.318	A	0.078	
59	rs1615413	F13B	intron 3	C/T	197030317	0.498	C	0.473	
60	rs6003	F13B	R115H exon3	A/G	197031021		G	0.078	
61	rs1412631	5' of F13B		C/T	197037237		T	0.078	mrd_3922
62	rs1794006	5' of F13B		C/T	197037449	0.348	T	0.079	rs1115246
63	rs6428380	5' of F13B		A/G	197037667	0.357	A	0.078	rs1115247

FIG. 2 CONT.

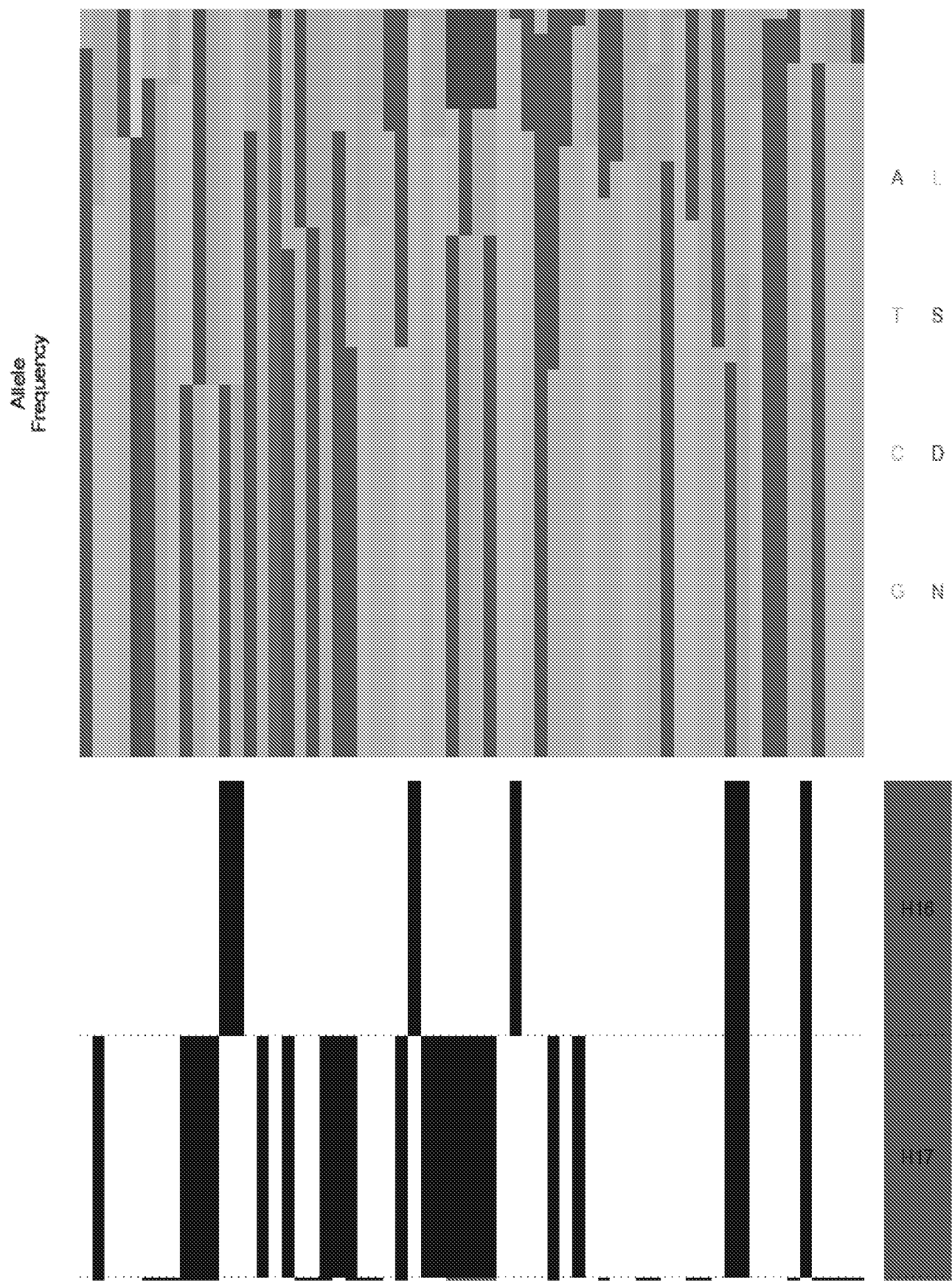


FIG. 3
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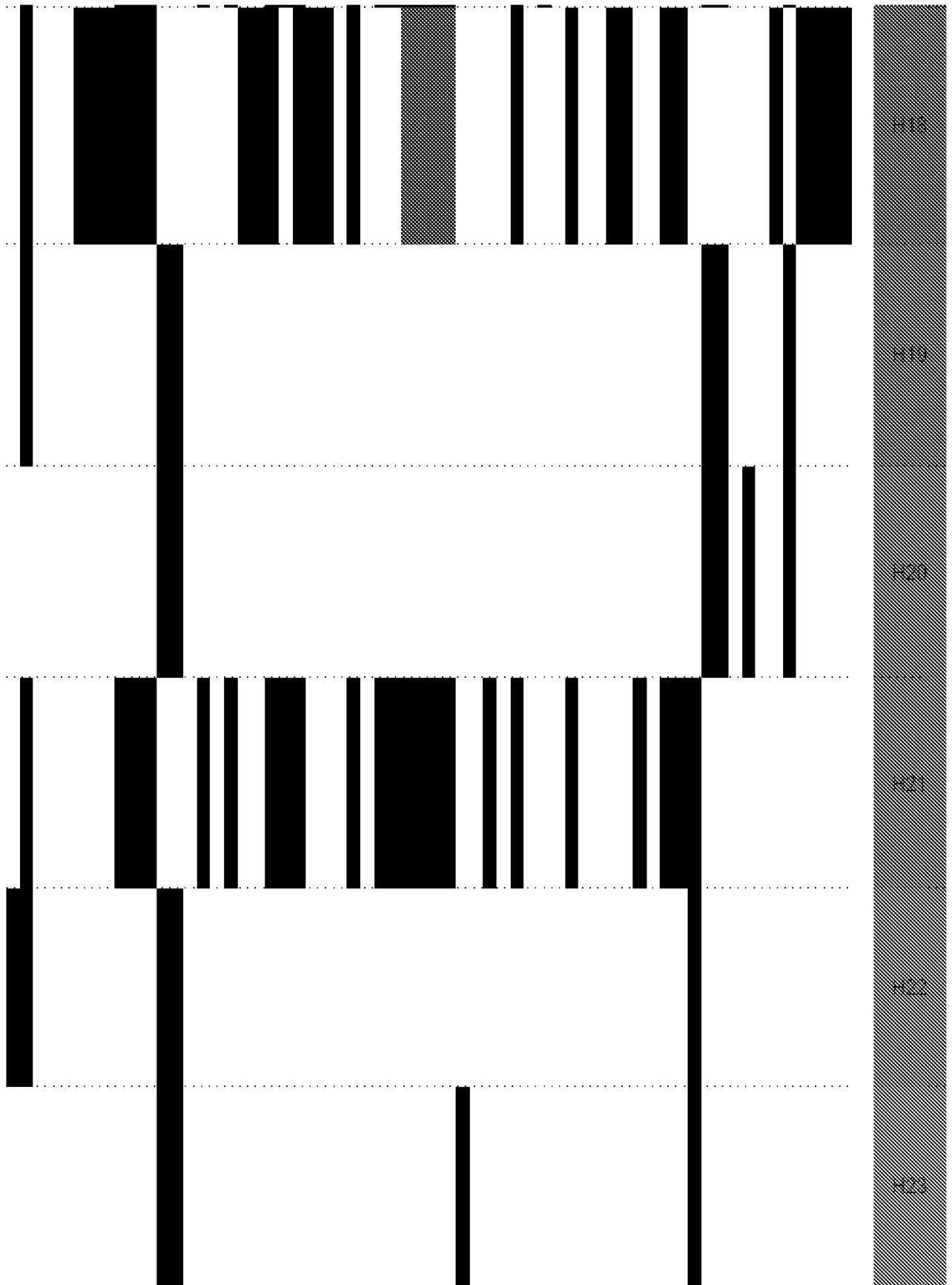


FIG. 3 CONT.

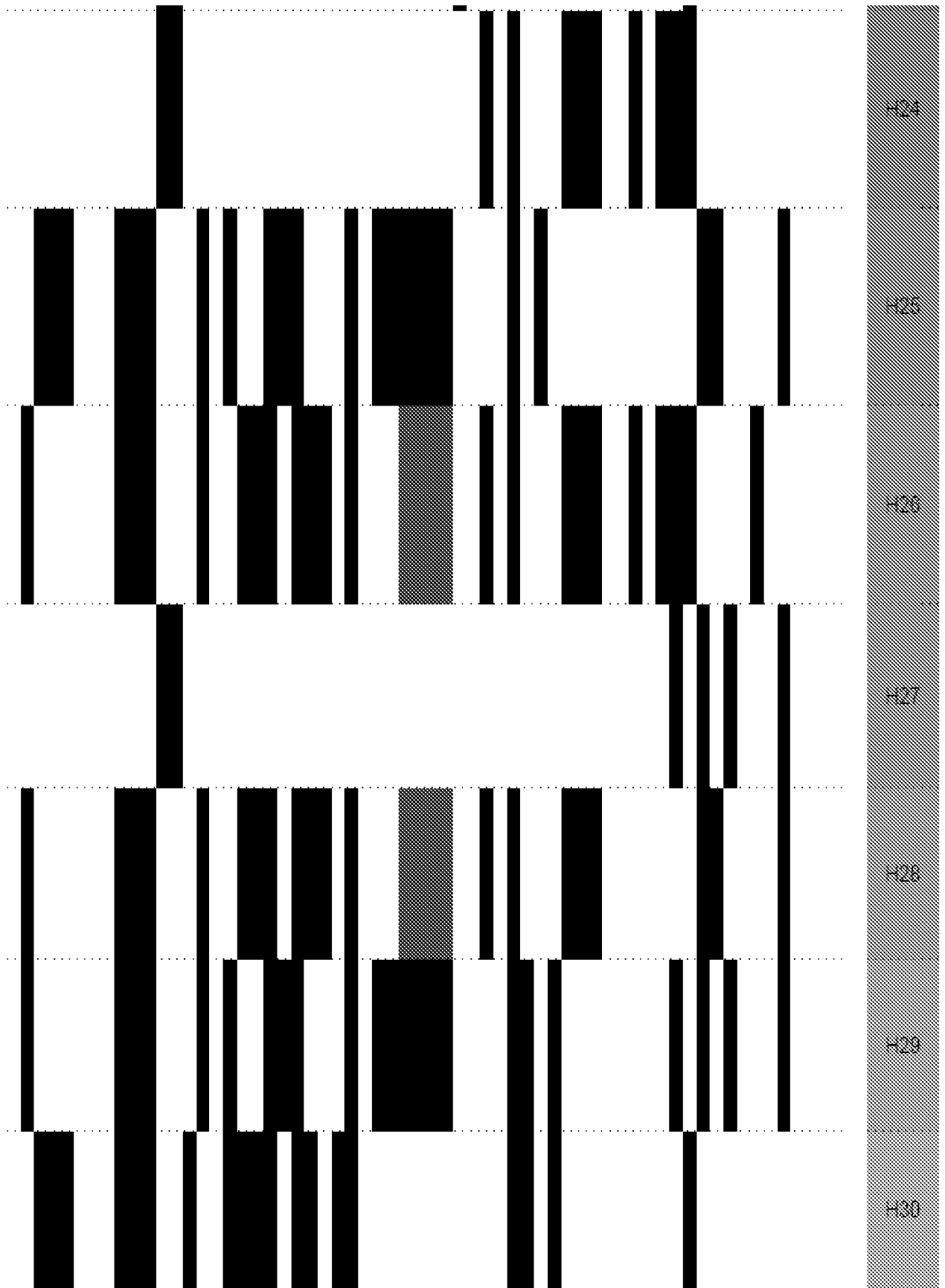


FIG. 3 CONT.

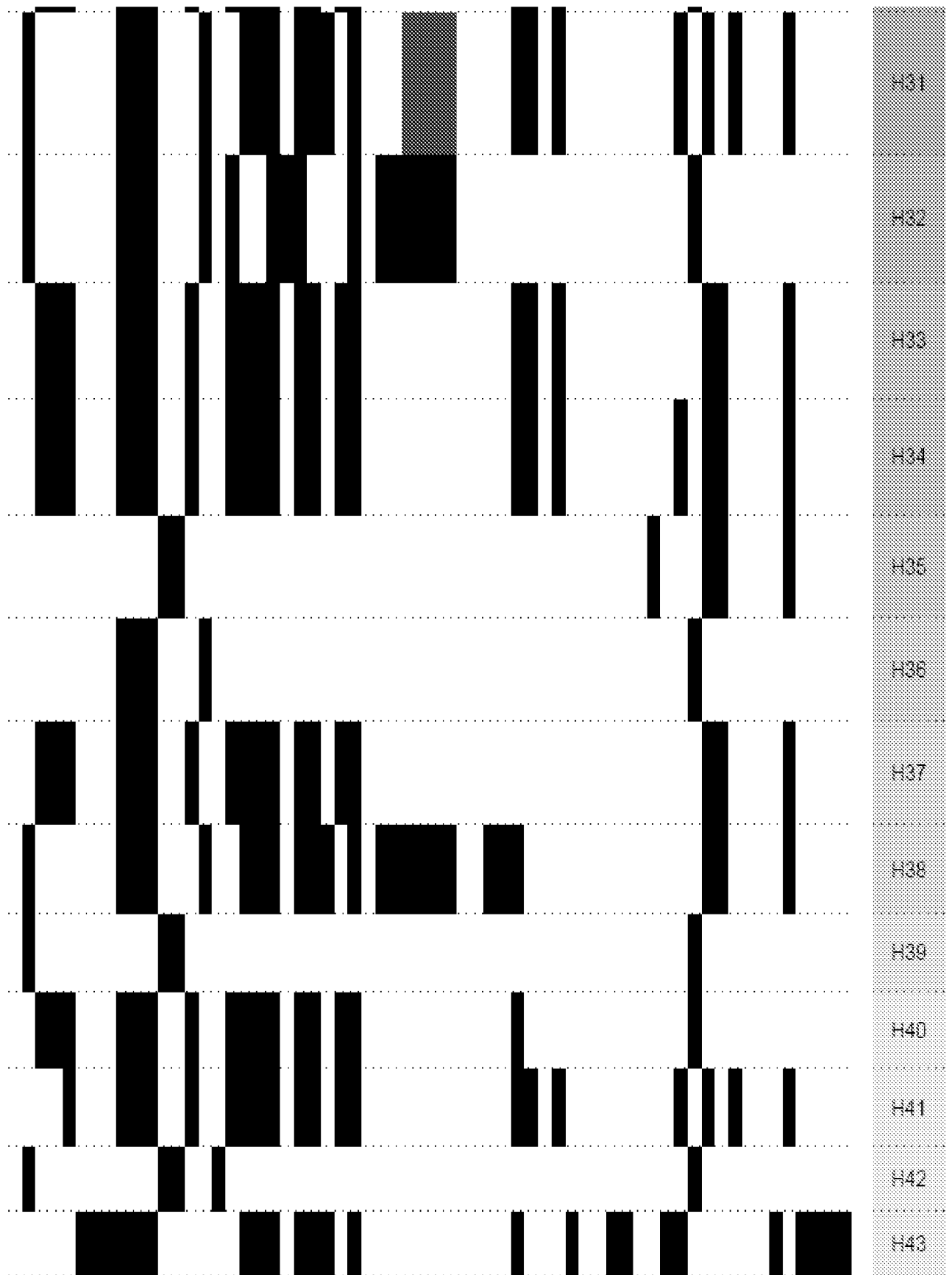


FIG. 3 CONT.

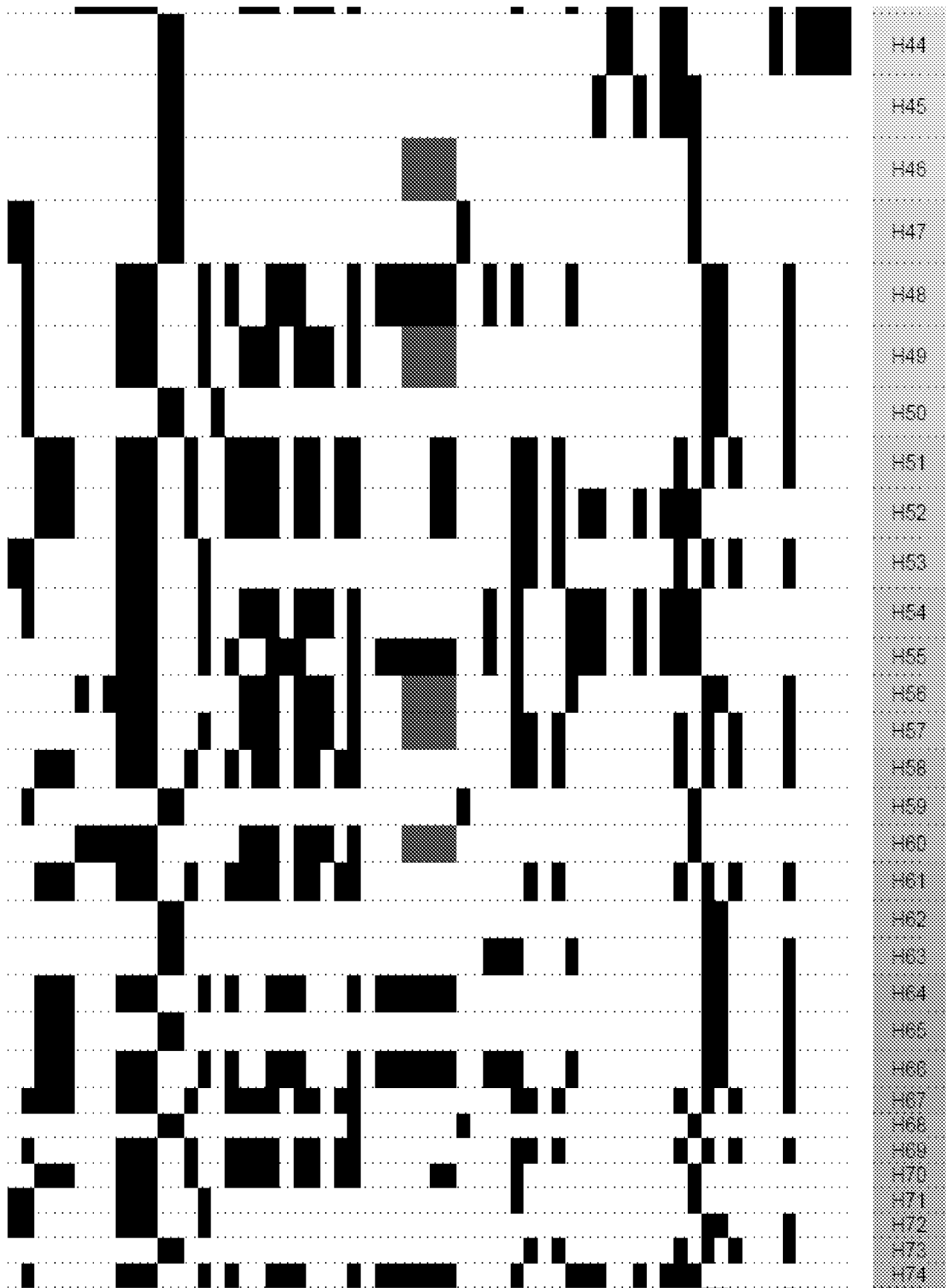


FIG. 3 CONT.

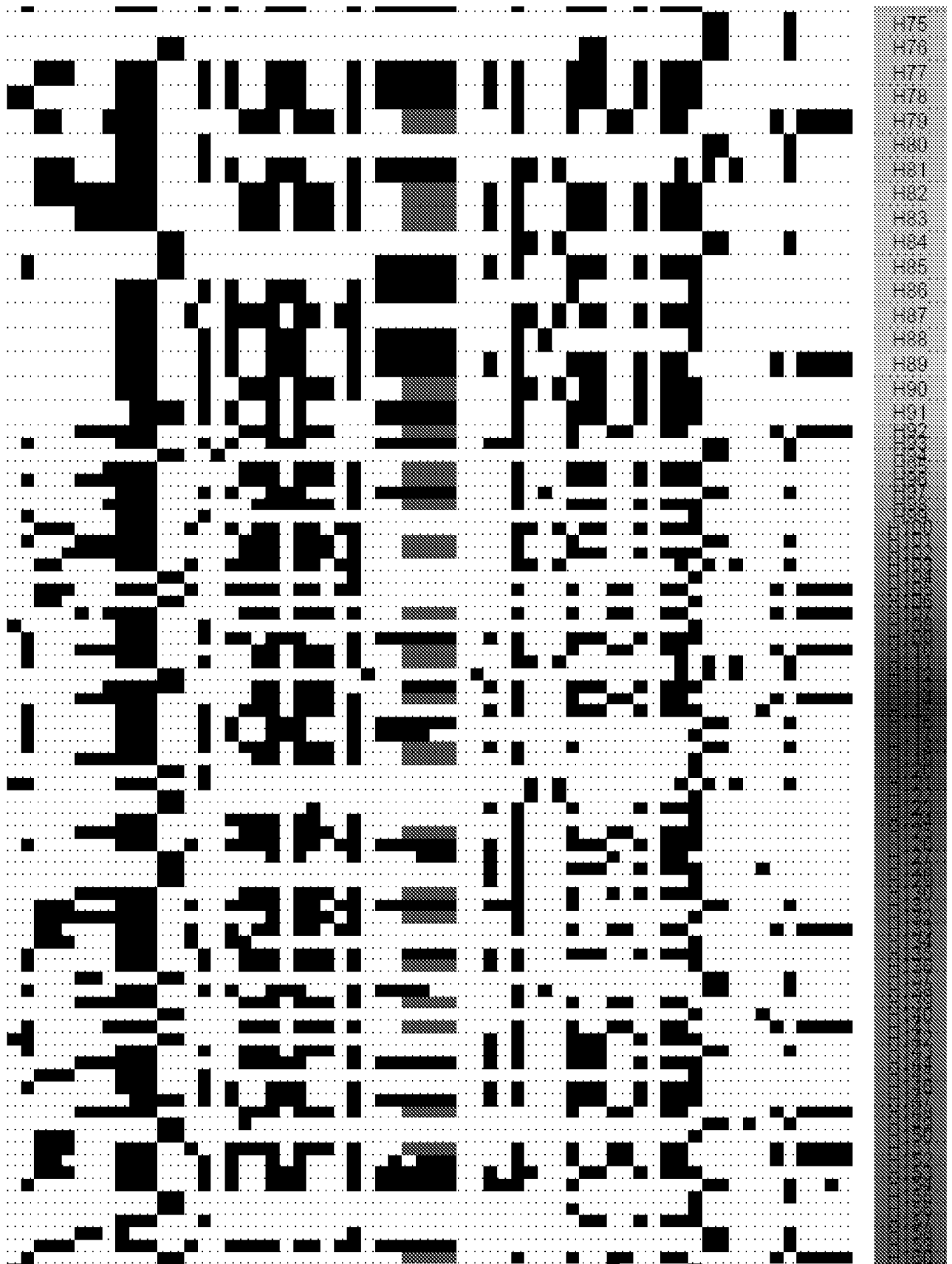
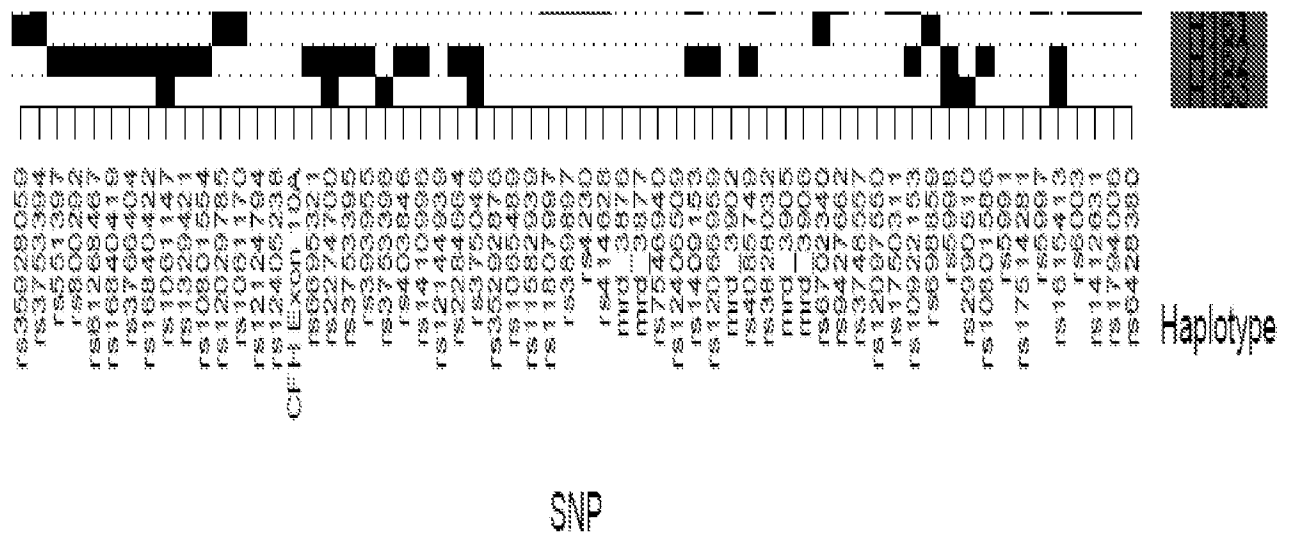


FIG. 3 CONT.



		GSH -1073 Haplotype	GSH 1073 sample Frequencies	GSH-927 Haplotype	GSH 927 sample Frequencies
H1	ACCGATCATCCAAGAGAACGGCGCCCGAGGGGGCGTACCGGGGCAAGCACG	H1	19.2%	H1	20.5%
H2	ACCGATCATCCAAGAGAACGGCGCCCGAGGGGGCGTACCGAATCAAGTACG	H2	15.4%	H2	16.7%
H3	ACTAATCCATTGATTAGTGACGCCCCGAAAGGGCGTACCTGGTTAAGCACG	H3	7.6%	H3	7.8%
H4	ATCGATCCATTAGGATGGCGGCTTCCAAAGGAATGTCATAATCAAGTACG	H4	6.6%	H4	6.3%
H5	ACCGGCTCATTAAATTAGTTGCGCCCGAAGGAGCTCACATGATCAAATGTA	H5	5.8%	H5	5.8%
H6	ACTAATCCATTGATTAGTGACGCCCCGAAAGGATGTCATAATCAAGTACG	H6	3.8%	H6	3.8%
H7	GTCGATCCATTAAAGAGAACGGCGCCCGAGGGGGCGTACCGAATCAAGTACG	H7	3.4%	H7	3.4%
H8	ACCGATCATCCAAGAGAACGGCGCCCGAGGGGATGTCATAATCAAGTACG	H8	2.1%	H8	2.4%
H9	ATCGATCCATTAGGATGGCGGCTTCCGAAGGGGCGTACCGGGGCAAGCACG	H9	2.0%	H9	1.9%
H10	ATCGATCCATTAGGATGGCGGCTTCCAGAGGAGCGTACCGGGGCAAGCACG	H10	1.9%	H11	1.7%
H11	ATCGATCCATTAAATTAGTTGCGCCCCAAGGAATGTCATAATCAAGTACG	H11	1.5%	H10	1.9%
H12	ACCGATCATCCAAGAGAACGGCGCCCGAAGGGCGTACCTGGTTAAGCACG	H12	1.4%	H12	1.5%
H13	ACCGGCTCATTAAATTAGTTGCGCCCGAAGGAATGTCATAATCAAGTACG	H13	1.4%	H14	1.2%
H14	ACCGATCATCCAAGAGAACGGCGCCCGAAGGGGCGTACCGGGGCAAGCACG	H14	1.3%	H13	1.2%
H15	ACCGGCTCATTAAATTAGTTGCGCCCGAAGGAGCGTACCGGGGCAAGCACG	H15	1.2%	H15	1.2%

FIG. 4

SNP	Gene	Allele	All Subjects				
			Odds Ratio	95% CI Lower	95% CI Upper	Chi Sq.	p value
rs35928059	CFH	G	0.986	0.640	1.520	0.004	0.9496
rs3753394	CFH	T	1.027	0.830	1.271	0.061	0.8043
rs551397	CFH	T	0.524	0.411	0.668	27.154	1.88E-07
rs800292	CFH	A	0.524	0.411	0.668	27.154	1.88E-07
rs61268467	CFH	G	0.512	0.402	0.653	29.276	6.28E-08
rs16840419	CFH	G	0.551	0.405	0.749	14.423	1.46E-04
rs3766404	CFH	C	0.546	0.399	0.747	14.294	0.0002
rs16840422	CFH	T	0.547	0.403	0.743	14.954	1.10E-04
rs1061147	CFH	A	1.966	1.615	2.394	45.387	1.62E-11
rs1329421	CFH	T	1.970	1.619	2.399	45.731	1.36E-11
rs10801554	CFH	C	1.970	1.619	2.399	45.731	1.36E-11
rs12029785	CFH	G	1.953	1.604	2.377	44.531	2.50E-11
rs1061170	CFH	C	1.953	1.604	2.377	44.531	2.50E-11
rs12124794	CFH	T	0.567	0.441	0.728	19.688	9.12E-06
rs12405238	CFH	T	0.855	0.692	1.056	2.125	0.1449
Exon10A	CFH	ins	2.217	0.468	10.501	1.007	0.3157
rs6695321	CFH	G	0.721	0.589	0.881	10.189	0.0014
rs2274700	CFH	A	0.434	0.351	0.535	60.451	7.54E-15
rs3753395	CFH	T	0.431	0.349	0.532	60.698	6.66E-15
rs393955	CFH	G	2.007	1.646	2.448	47.292	6.12E-12
rs3753396	CFH	G	1.092	0.840	1.418	0.429	0.5125
rs403846	CFH	A	2.007	1.646	2.448	47.292	6.12E-12
rs1410996	CFH	T	0.436	0.353	0.538	59.668	1.12E-14
rs12144939	CFH	T	0.454	0.348	0.592	33.825	6.03E-09
rs2284664	CFH	A	0.570	0.443	0.733	19.159	1.20E-05
rs375046	CFH	C	1.975	1.621	2.406	45.651	1.41E-11
rs35292876	CFH	T	2.796	0.803	9.729	2.611	0.1061
rs1065489	CFH	T	1.072	0.829	1.387	0.282	0.5954
rs11582939	CFH	T	1.072	0.829	1.387	0.282	0.5954
mrd_3876	CFHR2	G	0.977	0.427	2.237	0.003	0.9571
mrd_3877	CFHR2	A	2.796	0.803	9.729	2.611	0.1061

FIG. 5

rs7546940	CFHR4	A	0.883	0.685	1.138	0.922	0.3371
rs12406509	CFHR4	G	1.284	0.660	2.497	0.542	0.4616
rs1409153	CFHR4	G	1.940	1.594	2.360	43.770	3.69E-11
rs12066959	CFHR2	A	0.601	0.473	0.764	17.286	3.21E-05
mrd_3902	CFHR2	A	0.612	0.302	1.243	1.843	0.1746
rs4085749	CFHR2	T	0.606	0.477	0.771	16.665	4.46E-05
rs3828032	CFHR2	A	0.641	0.514	0.800	15.577	7.92E-05
mrd_3905	CFHR2	A	0.746	0.589	0.945	5.910	0.0151
mrd_3906	CFHR2	T	0.747	0.591	0.945	5.940	0.0148
rs6702340	CFHR5	T	0.485	0.342	0.688	16.448	5.00E-05
rs9427662	CFHR5	C	0.479	0.336	0.681	16.778	4.20E-05
rs3748557	CFHR5	T	0.729	0.576	0.922	6.983	0.0082
rs12097550	CFHR5	T	0.732	0.163	3.289	0.166	0.6838
rs1750311	CFHR5	A	0.592	0.478	0.734	22.826	1.77E-06
rs10922153	CFHR5	T	0.530	0.435	0.645	39.982	2.56E-10
rs698859	F13B	A	1.286	1.059	1.561	6.443	0.0111
rs5998	F13B	G	0.971	0.802	1.177	0.088	0.7662
rs2990510	F13B	G	1.201	0.981	1.470	3.144	0.0762
rs10801586	F13B	T	0.665	0.505	0.875	8.485	0.0036
rs5991	F13B	T	2.036	0.564	7.352	1.179	0.2776
rs17514281	F13B	G	0.421	0.155	1.142	2.889	0.0892
rs5997	F13B	A	0.479	0.336	0.681	16.778	4.20E-05
rs1615413	F13B	C	0.983	0.812	1.191	0.029	0.8645
rs6003	F13B	G	0.479	0.336	0.681	16.778	4.20E-05
rs1412631	F13B	T	0.479	0.336	0.681	16.778	4.20E-05
rs1794006	F13B	T	0.486	0.342	0.691	16.198	5.70E-05
rs6428380	F13B	A	0.479	0.336	0.681	16.778	4.20E-05
rs9332739	C2	C	0.466	0.286	0.760	9.357	0.0022
rs641153	CFB	T	0.662	0.483	0.906	6.629	0.0100
rs10490924	LOC387715/A	T	2.709	2.145	3.422	69.888	6.28E-17
rs11200638	HTRA1	A	2.538	2.024	3.183	65.139	6.98E-16
rs2230199	C3	G	1.384	1.101	1.739	7.755	0.0054

FIG. 5 CONT.

	case	non case	control	non control	Pearson Chi2	p-value	OR	lower 95% CI	upper 95% CI
H1_62_A	251	945	117	541	2.740	0.098	1.228	0.963	1.567
H2_62_A	230	966	61	597	31.820	<.0001	2.330	1.726	3.146
H3_62_A	67	1129	71	587	16.580	<.0001	0.491	0.346	0.695
H4_62_A	82	1114	34	624	2.060	0.151	1.351	0.895	2.039
H5_62_A	48	1148	53	605	13.460	0.000	0.477	0.319	0.714
H6_62_A	38	1158	29	629	1.840	0.175	0.712	0.435	1.165
H7_62_A	39	1157	23	635	0.070	0.791	0.931	0.551	1.572
H8_62_A	27	1169	15	643	0.000	1.000	0.990	0.523	1.875
H9_62_A	24	1172	11	647	0.260	0.610	1.205	0.586	2.475
H10_62_A	21	1175	9	649	0.400	0.527	1.289	0.587	2.831
H11_62_A	8	1188	21	637	17.540	<.0001	0.204	0.090	0.464
H12_62_A	19	1177	10	648	0.010	0.920	1.046	0.484	2.263
H13_62_A	9	1187	13	645	5.420	0.020	0.376	0.160	0.885
H14_62_A	15	1181	9	649	0.040	0.841	0.916	0.399	2.105
H15_62_A	14	1182	6	652	0.270	0.603	1.287	0.492	3.365

FIG. 6

Haplotype	rs35928059	rs800292	rs1061170	rs12144939	rs7546940	rs1409153	rs10922153	rs698859	AMD Association
Minor Allele	G	A	C	T	A	A	T	A	
MAF	5.08%	16.50%	48.97%	15.42%	15.38%	49.02%	40.87%	44.64%	
H1_62_A	A	G	C	G	G	G	G	G	Neutral
H2_62_A	A	G	C	G	G	G	G	A	Risk
H3_62_A	A	A	T	G	G	A	T	G	Protective
H4_62_A	A	G	T	G	A	A	T	A	Neutral
H5_62_A	A	G	T	T	G	A	T	G	Protective
H6_62_A	A	A	T	G	G	A	T	A	Neutral
H7_62_A	G	G	T	G	G	G	G	A	Neutral
H8_62_A	A	G	C	G	G	G	T	A	Neutral
H9_62_A	A	G	T	G	G	A	G	G	Neutral
H10_62_A	A	G	T	G	A	A	G	G	Neutral
H11_62_A	A	G	T	T	A	A	T	A	Protective
H12_62_A	A	G	C	G	G	A	T	G	Neutral
H13_62_A	A	G	T	T	G	A	T	A	Neutral
H14_62_A	A	G	C	G	G	A	G	G	Neutral
H15_62_A	A	G	T	T	G	A	G	G	Neutral

FIG. 7

Haplotype	rs35928059	rs800292	rs1061170	rs12144939	rs7546940	rs1409153	rs10922153	rs698859	AMD Association
Minor Allele	G	A	C	T	A	G	T	A	
H1_51_A	A	G	C	G	G	C	G	C	Neutral
H2_51_A	A	G	C	G	G	C	G	T	Risk
H3_51_A	A	A	T	G	G	T	T	C	Protective
H4_51_A	A	G	T	G	A	T	T	T	Neutral
H5_51_A	A	G	T	T	G	T	T	C	Protective
H6_51_A	A	A	T	G	G	T	T	T	Neutral
H7_51_A	G	G	T	G	G	C	G	T	Neutral
H8_51_A	A	G	C	G	G	C	T	T	Neutral
H9_51_A	A	G	T	G	G	T	G	C	Neutral
H10_51_A	A	G	T	T	A	T	T	T	Protective
H11_51_A	A	G	T	G	A	T	G	C	Neutral
H12_51_A	A	G	C	G	G	T	T	C	Neutral
H13_51_A	A	G	C	G	G	T	G	C	Neutral
H14_51_A	A	G	T	T	G	T	T	T	Neutral
H15_51_A	A	G	T	T	G	T	G	C	Neutral
H16_51_A	A	G	C	G	G	C	G	C	Neutral
H17_51_A	A	G	C	G	G	C	G	T	Neutral

FIG. 8

Haplotype	rs35928059	rs800292	rs1061170	rs12144939	rs7546940	rs1409153	rs10922153	rs698859	AMD Association
Minor Allele	G	A	C	T	A	C	T	T	
H1_51_B	A	G	C	G	G	C	G	C	Risk
H2_51_B	A	G	C	G	G	C	G	T	Risk
H3_51_B	A	A	T	G	G	T	T	C	Protective
H4_51_B	A	G	T	G	A	T	T	T	Neutral
H5_51_B	A	G	T	T	G	T	T	C	Protective
H6_51_B	A	A	T	G	G	T	T	T	Neutral
H7_51_B	G	G	T	G	G	C	G	T	Neutral
H8_51_B	A	G	C	G	G	C	T	T	Neutral
H9_51_B	A	G	T	G	A	T	G	C	Neutral
H10_51_B	A	G	C	G	G	C	G	C	Neutral
H11_51_B	A	G	T	G	G	T	G	C	Neutral
H12_51_B	A	G	T	T	A	T	T	T	Protective
H13_51_B	A	G	C	G	G	C	G	C	Neutral
H14_51_B	A	G	T	T	G	T	T	T	Protective
H15_51_B	A	G	T	T	G	T	G	C	Neutral
H16_51_B	A	G	T	T	G	T	T	C	Neutral
H17_51_B	A	G	C	G	G	T	T	C	Neutral
H18_51_B	A	G	T	G	G	T	T	C	Neutral
H19_51_B	A	G	C	G	G	T	G	C	Neutral
H20_51_B	A	G	C	G	A	T	T	T	Neutral
H21_51_B	A	G	T	G	G	T	G	C	Neutral
H22_51_B	A	G	C	G	G	C	G	C	Neutral

FIG. 9

H1 Haplotypes	62 SNPs	58 SNPs	51 SNPs	43 SNPs	31 SNPs	8 htSNPs
	P Value	P Value	P Value	P Value	P Value	P Value
All Subjects	0.116	0.1044	0.1375	0.0209	0.0179	0.0024
4B vs Control	0.0903	0.0992	0.1686			0.0049
4A vs control	0.6051	0.6051	0.5943			0.1369
Early AMD vs Control	0.0339	0.0394	0.0506			0.0094
H2 Haplotypes	62 SNPs	58 SNPs	51 SNPs	43 SNPs	31 SNPs	8 htSNPs
All Subjects	6.48E-09	1.55E-08	7.10E-09	2.58E-08	8.44E-09	1.13E-07
4B vs Control	1.09E-06	1.12E-06	1.45E-06			1.57E-05
4A vs control	8.35E-06	8.35E-06	3.92E-06			8.00E-04
Early AMD vs Control	1.12E-05	3.01E-05	1.05E-05			6.78E-07
H3 Haplotypes	62 SNPs	58 SNPs	51 SNPs	43 SNPs	31 SNPs	8 htSNPs
All Subjects	2.54E-05	1.24E-05	5.67E-05	9.89E-05	1.95E-05	6.46E-06
4B vs Control	4.00E-04	0.002	0.001			3.31E-05
4A vs control	0.132	0.132	0.1121			0.0551
Early AMD vs Control	4.00E-04	2.00E-04	6.00E-04			0.006
H5 Haplotypes	62 SNPs	58 SNPs	51 SNPs	43 SNPs	31 SNPs	8 htSNPs
All Subjects	1.00E-04	1.00E-04	7.78E-05	1.00E-04	3.00E-04	2.70E-06
4B vs Control	0.0021	0.0019	0.0019			4.00E-04
4A vs control	0.0858	0.0858	0.0418			0.0041
Early AMD vs Control	0.0077	0.0062	0.0059			4.00E-04
H11 Haplotypes	62 SNPs	58 SNPs	51 SNPs	43 SNPs	31 SNPs	8 htSNPs
All Subjects	8.80E-05	7.94E-05	7.14E-05	1.90E-05	4.00E-04	4.49E-05
4B vs Control	0.0019	0.0024	0.0012			2.00E-04
4A vs control	0.0369	0.0369	0.036			0.0266
Early AMD vs Control	0.0316	0.0277	0.0294			0.0489

FIG. 10

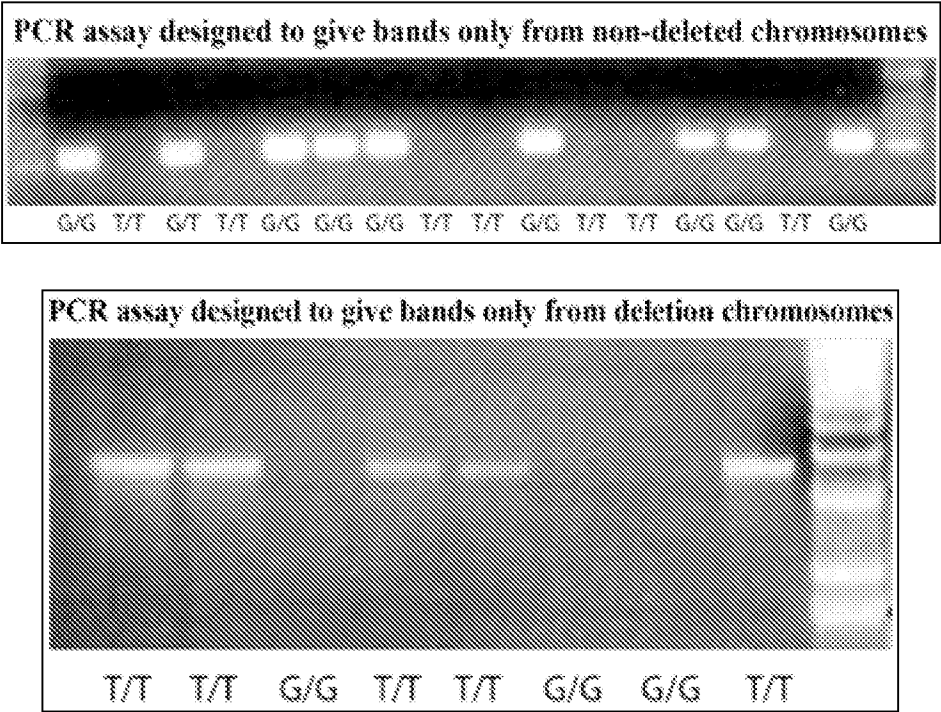


FIG. 11

Sibs - AIAMD v Norm	"Dominant" Model						"Additive" Model					
	Unadjusted						Unadjusted					
	Odds Ratio	95% CI Low	95% CI High	p value	Omnibus test		Odds Ratio	95% CI Low	95% CI High	p value	Omnibus test	
Sibsh1/GSHh1	1.531	0.829	2.826	0.1735			1.468	0.874	2.465	0.1466		
Sibsh2/GSHh2	2.563	1.308	5.020	0.0061			2.704	1.459	5.014	0.0016		
Sibsh3/GSHh3	0.602	0.277	1.312	0.2018			0.602	0.277	1.312	0.2018		
Sibsh4/GSHh4	0.666	0.291	1.522	0.3349			0.666	0.291	1.522	0.3349		
Sibsh5/GSHh5	0.147	0.040	0.536	0.0037			0.153	0.042	0.553	0.0042		
Sibsh6/GSHh7	1.184	0.446	3.145	0.7349			1.184	0.446	3.145	0.7349		
Sibsh7/GSHh6	0.486	0.167	1.414	0.1853			0.486	0.167	1.414	0.1853		
Sibsh8/GSHh16	1.075	0.297	3.892	0.9128			1.075	0.297	3.892	0.9128		
Sibsh9/GSHh10	1.166	0.344	3.953	0.8053			1.166	0.344	3.953	0.8053		
Sibsh10/GSHh9	0.834	0.227	3.066	0.7850			0.834	0.227	3.066	0.7850		
Sibsh11/GSHh11	0.725	0.166	3.172	0.6695			0.725	0.166	3.172	0.6695		
Sibsh12/GSHh12	1.000	0.258	3.870	1.0000			1.000	0.258	3.870	1.0000		
Sibsh13/GSHh13	0.640	0.085	4.797	0.6644			0.640	0.085	4.797	0.6644		
Sibsh15/GSHhX	3.646	0.371	35.835	0.2673			3.646	0.371	35.835	0.2673		
Sibsh16/GSHhX	0.194	0.020	1.892	0.1580			0.194	0.020	1.892	0.1580		
Sibsh17/GSHhX	1.000	0.141	7.099	1.0000			1.000	0.141	7.099	1.0000		
Sibsh18/GSHhX	1.155	0.190	7.032	0.8760			1.155	0.190	7.032	0.8760		
Sibsh19/GSHh8	1.319	0.217	8.009	0.7637	0.0356		1.319	0.217	8.009	0.7637	0.0131	

FIG. 12

Sibs - Neo v Norm	"Dominant" Model					"Additive" Model					
	Unadjusted					Unadjusted					
	Odds Ratio	95% CI Low	95% CI High	p value	Omnibus test	Odds Ratio	95% CI Low	95% CI High	p value	Omnibus test	
Sibsh1/GSHh1	1.673	0.873	3.207	0.1210		1.573	0.921	2.688	0.0974		
Sibsh2/GSHh2	2.587	1.286	5.207	0.0077		2.723	1.436	5.165	0.0022		
Sibsh3/GSHh3	0.589	0.271	1.280	0.1815		0.589	0.271	1.280	0.1815		
Sibsh4/GSHh4	0.608	0.255	1.450	0.2621		0.608	0.255	1.450	0.2621		
Sibsh5/GSHh5	0.110	0.024	0.497	0.0042		0.115	0.025	0.517	0.0048		
Sibsh6/GSHh7	1.180	0.415	3.360	0.7563		1.180	0.415	3.360	0.7563		
Sibsh7/GSHh6	0.488	0.149	1.595	0.2352		0.488	0.149	1.595	0.2352		
Sibsh8/GSHh16	1.075	0.297	3.892	0.9128		1.075	0.297	3.892	0.9128		
Sibsh9/GSHh10	0.640	0.164	2.492	0.5197		0.640	0.164	2.492	0.5197		
Sibsh10/GSHh9	0.838	0.232	3.031	0.7874		0.838	0.232	3.031	0.7874		
Sibsh11/GSHh11	0.798	0.186	3.429	0.7618		0.798	0.186	3.429	0.7618		
Sibsh12/GSHh12	1.000	0.229	4.373	1.0000		1.000	0.229	4.373	1.0000		
Sibsh13/GSHh13	0.640	0.085	4.797	0.6644		0.640	0.085	4.797	0.6644		
Sibsh15/GSHhX	3.000	0.312	28.841	0.3414		3.000	0.312	28.841	0.3414		
Sibsh16/GSHhX	0.229	0.023	2.255	0.2065		0.229	0.023	2.255	0.2065		
Sibsh17/GSHhX	1.000	0.141	7.099	1.0000		1.000	0.141	7.099	1.0000		
Sibsh18/GSHhX	1.686	0.232	12.232	0.6053		1.686	0.232	12.232	0.6053		
Sibsh19/GSHh8	1.000	0.141	7.099	1.0000		1.000	0.141	7.099	1.0000		0.0074

FIG. 12 CONT.

All nUP927 samples						
SNP No.	Name	Assoc. Allele	Case,Control Ratio Counts	Case,Control Freq.	Chi square	P value
1	rs2230199.C3	G	299:891, 126:530	0.251, 0.192	8.359	0.0038
2	rs9332739.C2	G	1165:31, 622:36	0.974, 0.945	10.102	0.0015
3	rs641153.CFB	C	1103:83, 582:70	0.930, 0.893	7.703	0.0055
4	rs35928059	A	1135:61, 624:34	0.949, 0.948	0.004	0.9502
5	rs3753394	T	310:886, 167:491	0.259, 0.254	0.065	0.7992
6	rs551397	C	1043:153, 508:150	0.872, 0.772	31.07	2.49E-08
7	rs800292	G	1043:153, 508:150	0.872, 0.772	31.07	2.49E-08
8	rs61268467	A	1043:153, 505:153	0.872, 0.767	33.698	6.44E-09
9	rs16840419	A	1098:98, 567:91	0.918, 0.862	14.727	1.00E-04
10	rs3766404	T	1100:96, 569:89	0.920, 0.865	14.29	2.00E-04
11	rs16840422	C	1098:98, 566:92	0.918, 0.860	15.459	8.43E-05
12	rs1061147	A	678:518, 261:397	0.567, 0.397	49.212	2.30E-12
13	rs1329421	T	679:517, 261:397	0.568, 0.397	49.698	1.79E-12
14	rs10801554	C	679:517, 261:397	0.568, 0.397	49.698	1.79E-12
15	rs12029785	G	676:520, 261:397	0.565, 0.397	48.247	3.76E-12
16	rs1061170	C	676:520, 261:397	0.565, 0.397	48.247	3.76E-12
17	rs12124794	A	1050:146, 524:134	0.878, 0.796	22.03	2.68E-06
18	rs12405238	G	923:273, 487:171	0.772, 0.740	2.33	0.1269
19	CFH_Exon_101	G	8:1188, 2:656	0.007, 0.003	1.054	0.3046
20	rs6695321	A	869:327, 430:228	0.727, 0.653	10.812	0.001
21	rs2274700	G	920:276, 386:272	0.769, 0.587	67.978	1.65E-16
22	rs3753395	A	916:280, 384:274	0.766, 0.584	67.326	2.30E-16
23	rs393955	G	734:462, 291:367	0.614, 0.442	50.481	1.20E-12
24	rs3753396	G	183:1013, 93:565	0.153, 0.141	0.456	0.4993
25	rs403846	A	734:462, 291:367	0.614, 0.442	50.481	1.20E-12
26	rs1410996	C	915:281, 384:274	0.765, 0.584	66.641	3.26E-16
27	rs12144939	G	1063:133, 517:141	0.889, 0.786	35.811	2.17E-09
28	rs2284664	G	1050:146, 525:133	0.878, 0.798	21.279	3.97E-06
29	rs375046	C	731:465, 291:367	0.611, 0.442	48.982	2.58E-12
30	rs35292876	T	15:1181, 3:655	0.013, 0.005	2.813	0.0935
31	rs1065489	T	186:1010, 96:562	0.156, 0.146	0.305	0.581
32	rs11582939	T	186:1010, 96:562	0.156, 0.146	0.305	0.581
33	rs11807997 Deletion	A	881:187, 425:96	0.825, 0.816	0.201	0.6539

FIG. 13A

32	rs11582939	T	186:1010, 96:562	0.156, 0.146	0.305	0.581
33	rs11807997 Deletion	A	881:187, 425:96	0.825, 0.816	0.201	0.6539
34	rs389897 Deletion	T	880:188, 425:96	0.824, 0.816	0.162	0.6877
35	rs4230 Deletion	G	875:193, 423:98	0.819, 0.812	0.128	0.7208
36	rs414628 Deletion	A	875:193, 423:98	0.819, 0.812	0.128	0.7208
37	mrd_3876	C	1180:16, 649:9	0.987, 0.986	0.003	0.9573
38	mrd_3877	A	15:1181, 3:655	0.013, 0.005	2.813	0.0935
39	rs7546940	G	1024:172, 552:106	0.856, 0.839	0.995	0.3186
40	rs12406509	G	30:1166, 13:645	0.025, 0.020	0.532	0.4659
41	rs1409153	G	699:497, 275:383	0.584, 0.418	47.199	6.41E-12
42	rs12066959	G	1024:172, 511:147	0.856, 0.777	18.876	1.40E-05
43	mrd_3902	G	1179:17, 643:15	0.986, 0.977	1.843	0.1746
44	rs4085749	C	1024:172, 512:146	0.856, 0.778	18.207	1.98E-05
45	rs3828032	G	935:261, 460:198	0.782, 0.699	15.579	7.91E-05
46	mrd_3905	G	985:211, 511:147	0.824, 0.777	6.014	0.0142
47	mrd_3906	C	982:214, 509:149	0.821, 0.774	6.086	0.0136
48	rs6702340	G	1127:69, 585:73	0.942, 0.889	17.018	3.70E-05
49	rs9427662	T	1129:67, 586:72	0.944, 0.891	17.455	2.94E-05
50	rs3748557	A	983:213, 507:151	0.822, 0.771	7.104	0.0077
51	rs12097550	C	1192:4, 655:3	0.997, 0.995	0.167	0.6832
52	rs1750311	C	915:281, 435:223	0.765, 0.661	23.174	1.48E-06
53	rs10922153	G	786:410, 330:328	0.657, 0.502	42.93	5.67E-11
54	rs698859	A	564:632, 270:388	0.472, 0.410	6.432	0.0112
55	rs5998	A	632:564, 343:315	0.528, 0.521	0.087	0.7679
56	rs2990510	G	438:758, 214:444	0.366, 0.325	3.128	0.0769
57	rs10801586	C	1070:126, 557:101	0.895, 0.847	9.157	0.0025
58	rs5991	T	11:1185, 3:655	0.009, 0.005	1.218	0.2697
59	rs17514281	A	1189:7, 649:9	0.994, 0.986	3.038	0.0813
60	rs5997	G	1129:67, 586:72	0.944, 0.891	17.455	2.94E-05
61	rs1615413	T	632:564, 345:313	0.528, 0.524	0.029	0.8653
62	rs6003	A	1129:67, 586:72	0.944, 0.891	17.455	2.94E-05
63	rs1412631	C	1129:67, 586:72	0.944, 0.891	17.455	2.94E-05
64	rs1794006	C	1128:68, 586:72	0.943, 0.891	16.801	4.15E-05
65	rs6428380	G	1129:67, 586:72	0.944, 0.891	17.455	2.94E-05

FIG. 13A CONT.

nUP927, all SNPs, all Loci				
SNP No.	Name	P value All	P value Female	P value Male
1	rs2230199.C3	0.0038	0.0092	0.2317
2	rs9332739.C2	0.0015	0.0089	0.0964
3	rs641153.CFB	0.0055	0.0407	0.0853
4	rs35928059	0.9502	0.5279	0.5223
5	rs3753394	0.7992	0.8554	0.845
6	rs551397	2.49E-08	1.92E-10	0.2421
7	rs800292	2.49E-08	1.92E-10	0.2421
8	rs61268467	6.44E-09	7.67E-11	0.1719
9	rs16840419	1.00E-04	2.00E-04	0.1385
10	rs3766404	2.00E-04	9.70E-05	0.2238
11	rs16840422	8.43E-05	1.00E-04	0.1385
12	rs1061147	2.30E-12	1.81E-13	0.0451
13	rs1329421	1.79E-12	1.32E-13	0.0451
14	rs10801554	1.79E-12	1.32E-13	0.0451
15	rs12029785	3.76E-12	3.37E-13	0.0451
16	rs1061170	3.76E-12	3.37E-13	0.0451
17	rs12124794	2.68E-06	2.71E-07	0.2099
18	rs12405238	0.1269	0.073	0.91
19	CFH_Exon_101	0.3046	0.3162	0.792
20	rs6695321	0.001	3.86E-06	0.7976
21	rs2274700	1.65E-16	7.48E-13	3.27E-05
22	rs3753395	2.30E-16	2.90E-13	7.94E-05
23	rs393955	1.20E-12	5.90E-13	0.0158
24	rs3753396	0.4993	0.4604	0.0659
25	rs403846	1.20E-12	5.90E-13	0.0158
26	rs1410996	3.26E-16	6.31E-13	5.94E-05
27	rs12144939	2.17E-09	6.38E-06	2.00E-04
28	rs2284664	3.97E-06	8.63E-07	0.1704
29	rs375046	2.58E-12	1.52E-12	0.0158
30	rs35292876	0.0935	0.6069	0.0412
31	rs1065489	0.581	0.3277	0.0551
32	rs11582939	0.581	0.3277	0.0551
33	rs11807997 Deletion	0.6539	0.0697	0.162

FIG. 13B

34	rs389897 Deletion	0.6877	0.0697	0.14
35	rs4230 Deletion	0.7208	0.0757	0.14
36	rs414628 Deletion	0.7208	0.0757	0.14
37	mrd_3876	0.9573	0.4563	0.2093
38	mrd_3877	0.0935	0.6069	0.0412
39	rs7546940	0.3186	0.2715	0.935
40	rs12406509	0.4659	0.8802	0.3192
41	rs1409153	6.41E-12	2.78E-13	0.0526
42	rs12066959	1.40E-05	1.25E-05	0.1121
43	mrd_3902	0.1746	0.0141	0.4579
44	rs4085749	1.98E-05	1.25E-05	0.1395
45	rs3828032	7.91E-05	4.49E-05	0.2408
46	mrd_3905	0.0142	0.0199	0.3666
47	mrd_3906	0.0136	0.0235	0.3131
48	rs6702340	3.70E-05	2.74E-05	0.1378
49	rs9427662	2.94E-05	3.31E-05	0.1073
50	rs3748557	0.0077	0.0111	0.3131
51	rs12097550	0.6832	0.1763	0.2397
52	rs1750311	1.48E-06	2.22E-06	0.0745
53	rs10922153	5.67E-11	1.54E-11	0.0333
54	rs698859	0.0112	0.0349	0.1227
55	rs5998	0.7679	0.9358	0.5097
56	rs2990510	0.0769	0.0139	0.8079
57	rs10801586	0.0025	5.00E-04	0.5134
58	rs5991	0.2697	0.3162	0.5084
59	rs17514281	0.0813	0.2698	0.2093
60	rs5997	2.94E-05	6.18E-05	0.078
61	rs1615413	0.8653	0.9686	0.673
62	rs6003	2.94E-05	6.18E-05	0.078
63	rs1412631	2.94E-05	6.18E-05	0.078
64	rs1794006	4.15E-05	6.18E-05	0.1018
65	rs6428380	2.94E-05	6.18E-05	0.078

FIG. 13B CONT.

Female nUP927 samples						
SNP No.	Name	Assoc Allele	Case,Control Ratio Counts	Case,Control Freq.	Chi square	P value
1	rs2230199.C3	G	199:551, 68:284	0.265, 0.193	6.793	0.0092
2	rs9332739.C2	G	737:15, 335:17	0.980, 0.952	6.846	0.0089
3	rs641153.CFB	C	697:49, 311:35	0.934, 0.899	4.189	0.0407
4	rs35928059	A	716:36, 332:20	0.952, 0.943	0.398	0.5279
5	rs3753394	T	194:558, 89:263	0.258, 0.253	0.033	0.8554
6	rs551397	C	670:82, 261:91	0.891, 0.741	40.542	1.92E-10
7	rs800292	G	670:82, 261:91	0.891, 0.741	40.542	1.92E-10
8	rs61268467	A	669:83, 259:93	0.890, 0.736	42.341	7.67E-11
9	rs16840419	A	698:54, 302:50	0.928, 0.858	13.862	2.00E-04
10	rs3766404	T	700:52, 302:50	0.931, 0.858	15.194	9.70E-05
11	rs16840422	C	698:54, 301:51	0.928, 0.855	14.878	1.00E-04
12	rs1061147	A	452:300, 128:224	0.601, 0.364	54.204	1.81E-13
13	rs1329421	T	453:299, 128:224	0.602, 0.364	54.823	1.32E-13
14	rs10801554	C	453:299, 128:224	0.602, 0.364	54.823	1.32E-13
15	rs12029785	G	450:302, 128:224	0.598, 0.364	52.978	3.37E-13
16	rs1061170	C	450:302, 128:224	0.598, 0.364	52.978	3.37E-13
17	rs12124794	A	668:84, 271:81	0.888, 0.770	26.446	2.71E-07
18	rs12405238	G	592:160, 260:92	0.787, 0.739	3.215	0.073
19	CFH_Exon_101	G	6:746, 1:351	0.008, 0.003	1.005	0.3162
20	rs6695321	A	558:194, 213:139	0.742, 0.605	21.335	3.86E-06
21	rs2274700	G	592:160, 204:148	0.787, 0.580	51.415	7.48E-13
22	rs3753395	A	591:161, 202:150	0.786, 0.574	53.276	2.90E-13
23	rs393955	G	481:271, 144:208	0.640, 0.409	51.879	5.90E-13
24	rs3753396	A	641:111, 294:58	0.852, 0.835	0.545	0.4604
25	rs403846	A	481:271, 144:208	0.640, 0.409	51.879	5.90E-13
26	rs1410996	C	589:163, 202:150	0.783, 0.574	51.747	6.31E-13
27	rs12144939	G	675:77, 281:71	0.898, 0.798	20.371	6.38E-06
28	rs2284664	G	668:84, 273:79	0.888, 0.776	24.212	8.63E-07
29	rs375046	C	478:274, 144:208	0.636, 0.409	50.028	1.52E-12
30	rs35292876	T	9:743, 3:349	0.012, 0.009	0.265	0.6069
31	rs1065489	G	639:113, 291:61	0.850, 0.827	0.958	0.3277
32	rs11582939	C	639:113, 291:61	0.850, 0.827	0.958	0.3277
33	rs11807997	A	567:113, 222:61	0.834, 0.784	3.29	0.0697

FIG. 13C

34	rs389897	T	567:113, 222:61	0.834, 0.784	3.29	0.0697
35	rs4230	G	562:118, 220:63	0.826, 0.777	3.155	0.0757
36	rs414628	A	562:118, 220:63	0.826, 0.777	3.155	0.0757
37	mrd_3876	G	13:739, 4:348	0.017, 0.011	0.555	0.4563
38	mrd_3877	A	9:743, 3:349	0.012, 0.009	0.265	0.6069
39	rs7546940	G	653:99, 297:55	0.868, 0.844	1.209	0.2715
40	rs12406509	G	16:736, 7:345	0.021, 0.020	0.023	0.8802
41	rs1409153	G	461:291, 133:219	0.613, 0.378	53.36	2.78E-13
42	rs12066959	G	650:102, 267:85	0.864, 0.759	19.09	1.25E-05
43	mrd_3902	G	744:8, 341:11	0.989, 0.969	6.022	0.0141
44	rs4085749	C	650:102, 267:85	0.864, 0.759	19.09	1.25E-05
45	rs3828032	G	603:149, 243:109	0.802, 0.690	16.651	4.49E-05
46	mrd_3905	G	629:123, 274:78	0.836, 0.778	5.421	0.0199
47	mrd_3906	C	626:126, 273:79	0.832, 0.776	5.13	0.0235
48	rs6702340	G	712:40, 308:44	0.947, 0.875	17.587	2.74E-05
49	rs9427662	T	713:39, 309:43	0.948, 0.878	17.232	3.31E-05
50	rs3748557	A	627:125, 271:81	0.834, 0.770	6.448	0.0111
51	rs12097550	C	750:2, 349:3	0.997, 0.991	1.828	0.1763
52	rs1750311	C	588:164, 228:124	0.782, 0.648	22.391	2.22E-06
53	rs10922153	G	512:240, 165:187	0.681, 0.469	45.478	1.54E-11
54	rs698859	A	350:402, 140:212	0.465, 0.398	4.452	0.0349
55	rs5998	G	363:389, 169:183	0.483, 0.480	0.006	0.9358
56	rs2990510	G	288:464, 108:244	0.383, 0.307	6.046	0.0139
57	rs10801586	C	677:75, 291:61	0.900, 0.827	12.012	5.00E-04
58	rs5991	T	6:746, 1:351	0.008, 0.003	1.005	0.3162
59	rs17514281	A	748:4, 348:4	0.995, 0.989	1.218	0.2698
60	rs5997	G	713:39, 310:42	0.948, 0.881	16.048	6.18E-05
61	rs1615413	C	362:390, 169:183	0.481, 0.480	0.002	0.9686
62	rs6003	A	713:39, 310:42	0.948, 0.881	16.048	6.18E-05
63	rs1412631	C	713:39, 310:42	0.948, 0.881	16.048	6.18E-05
64	rs1794006	C	713:39, 310:42	0.948, 0.881	16.048	6.18E-05
65	rs6428380	G	713:39, 310:42	0.948, 0.881	16.048	6.18E-05

FIG. 13C CONT.

Male nUP927 samples						
SNP No.	Name	Assoc Allele	Case,Control Ratio Counts	Case,Control Freq.	Chi square	P value
1	rs2230199.C3	G	100:340, 58:246	0.227, 0.191	1.431	0.2317
2	rs9332739.C2	G	428:16, 287:19	0.964, 0.938	2.764	0.0964
3	rs641153.CFB	C	406:34, 271:35	0.923, 0.886	2.961	0.0853
4	rs35928059	G	25:419, 14:292	0.056, 0.046	0.409	0.5223
5	rs3753394	T	116:328, 78:228	0.261, 0.255	0.038	0.845
6	rs551397	C	373:71, 247:59	0.840, 0.807	1.368	0.2421
7	rs800292	G	373:71, 247:59	0.840, 0.807	1.368	0.2421
8	rs61268467	A	374:70, 246:60	0.842, 0.804	1.866	0.1719
9	rs16840419	A	400:44, 265:41	0.901, 0.866	2.194	0.1385
10	rs3766404	T	400:44, 267:39	0.901, 0.873	1.48	0.2238
11	rs16840422	C	400:44, 265:41	0.901, 0.866	2.194	0.1385
12	rs1061147	A	226:218, 133:173	0.509, 0.435	4.015	0.0451
13	rs1329421	T	226:218, 133:173	0.509, 0.435	4.015	0.0451
14	rs10801554	C	226:218, 133:173	0.509, 0.435	4.015	0.0451
15	rs12029785	G	226:218, 133:173	0.509, 0.435	4.015	0.0451
16	rs1061170	C	226:218, 133:173	0.509, 0.435	4.015	0.0451
17	rs12124794	A	382:62, 253:53	0.860, 0.827	1.572	0.2099
18	rs12405238	G	331:113, 227:79	0.745, 0.742	0.013	0.91
19	CFH_Exon_101	G	2:442, 1:305	0.005, 0.003	0.07	0.792
20	rs6695321	G	133:311, 89:217	0.300, 0.291	0.066	0.7976
21	rs2274700	G	328:116, 182:124	0.739, 0.595	17.255	3.27E-05
22	rs3753395	A	325:119, 182:124	0.732, 0.595	15.571	7.94E-05
23	rs393955	G	253:191, 147:159	0.570, 0.480	5.821	0.0158
24	rs3753396	G	72:372, 35:271	0.162, 0.114	3.382	0.0659
25	rs403846	A	253:191, 147:159	0.570, 0.480	5.821	0.0158
26	rs1410996	C	326:118, 182:124	0.734, 0.595	16.121	5.94E-05
27	rs12144939	G	388:56, 236:70	0.874, 0.771	13.651	2.00E-04
28	rs2284664	G	382:62, 252:54	0.860, 0.824	1.88	0.1704
29	rs375046	C	253:191, 147:159	0.570, 0.480	5.821	0.0158
30	rs35292876	T	6:438, 0:306	0.014, 0.000	4.168	0.0412
31	rs1065489	T	73:371, 35:271	0.164, 0.114	3.679	0.0551
32	rs11582939	T	73:371, 35:271	0.164, 0.114	3.679	0.0551
33	rs11807997	G	74:314, 35:203	0.191, 0.147	1.956	0.162

FIG. 13D

34	rs389897	A	75:313, 35:203	0.193, 0.147	2.178	0.14
35	rs4230	T	75:313, 35:203	0.193, 0.147	2.178	0.14
36	rs414628	T	75:313, 35:203	0.193, 0.147	2.178	0.14
37	mrd_3876	C	441:3, 301:5	0.993, 0.984	1.576	0.2093
38	mrd_3877	A	6:438, 0:306	0.014, 0.000	4.168	0.0412
39	rs7546940	G	371:73, 255:51	0.836, 0.833	0.007	0.935
40	rs12406509	G	14:430, 6:300	0.032, 0.020	0.992	0.3192
41	rs1409153	G	238:206, 142:164	0.536, 0.464	3.755	0.0526
42	rs12066959	G	374:70, 244:62	0.842, 0.797	2.525	0.1121
43	mrd_3902	A	9:435, 4:302	0.020, 0.013	0.551	0.4579
44	rs4085749	C	374:70, 245:61	0.842, 0.801	2.184	0.1395
45	rs3828032	G	332:112, 217:89	0.748, 0.709	1.376	0.2408
46	mrd_3905	G	356:88, 237:69	0.802, 0.775	0.815	0.3666
47	mrd_3906	C	356:88, 236:70	0.802, 0.771	1.017	0.3131
48	rs6702340	G	415:29, 277:29	0.935, 0.905	2.203	0.1378
49	rs9427662	T	416:28, 277:29	0.937, 0.905	2.594	0.1073
50	rs3748557	A	356:88, 236:70	0.802, 0.771	1.017	0.3131
51	rs12097550	T	2:442, 0:306	0.005, 0.000	1.382	0.2397
52	rs1750311	C	327:117, 207:99	0.736, 0.676	3.182	0.0745
53	rs10922153	G	274:170, 165:141	0.617, 0.539	4.529	0.0333
54	rs698859	A	214:230, 130:176	0.482, 0.425	2.383	0.1227
55	rs5998	A	243:201, 160:146	0.547, 0.523	0.435	0.5097
56	rs2990510	T	294:150, 200:106	0.662, 0.654	0.059	0.8079
57	rs10801586	C	393:51, 266:40	0.885, 0.869	0.427	0.5134
58	rs5991	T	5:439, 2:304	0.011, 0.007	0.437	0.5084
59	rs17514281	A	441:3, 301:5	0.993, 0.984	1.576	0.2093
60	rs5997	G	416:28, 276:30	0.937, 0.902	3.106	0.078
61	rs1615413	T	242:202, 162:144	0.545, 0.529	0.178	0.673
62	rs6003	A	416:28, 276:30	0.937, 0.902	3.106	0.078
63	rs1412631	C	416:28, 276:30	0.937, 0.902	3.106	0.078
64	rs1794006	C	415:29, 276:30	0.935, 0.902	2.676	0.1018
65	rs6428380	G	416:28, 276:30	0.937, 0.902	3.106	0.078

FIG. 13D CONT.

SNP No.	Name	Position	ObsHET	PredHET	HWpval	%Geno	FamTrio	MendErr	MAF	Alleles
1	rs17700121	194812498	0.233	0.231	1	100	30	0	0.133	A:T
2	rs4603077	194812514	0	0	1	100	30	0	0	T:T
3	rs4294368	194814311	0.125	0.117	1	90	22	0	0.062	T:C
4	rs4518841	194814498	0.133	0.124	1	100	30	0	0.067	G:A
5	rs7544266	194814552	0.102	0.097	1	98.9	29	0	0.051	A:G
6	rs12726412	194814597	0.05	0.049	1	100	30	0	0.025	T:C
7	rs4598443	194817209	0	0	1	88.9	25	0	0	A:A
8	rs12757236	194817578	0.034	0.033	1	98.9	29	0	0.017	C:A
9	rs16840189	194817841	0	0	1	100	30	0	0	C:C
10	rs12727032	194818726	0.433	0.444	1	100	30	0	0.333	A:G
11	rs10922089	194819744	0	0	1	100	30	0	0	A:A
12	rs6702610	194819980	0.086	0.082	1	97.8	28	0	0.043	C:T
13	rs12130043	194820206	0	0	1	100	30	0	0	A:A
14	rs4658041	194820376	0	0	1	98.9	29	0	0	C:C
15	rs4658042	194821025	0.133	0.124	1	100	30	0	0.067	G:T
16	rs12034150	194821643	0.424	0.443	0.9123	98.9	29	0	0.331	A:G
17	rs10737676	194822492	0.127	0.119	1	93.3	25	0	0.064	C:G
18	rs12127468	194822576	0	0	1	100	30	0	0	T:T
19	rs10737677	194822866	0	0	1	100	30	0	0	G:G
20	rs10737678	194822922	0.119	0.112	1	98.9	29	0	0.059	A:G
21	rs6703584	194823267	0	0	1	98.9	29	0	0	G:G
22	rs12128544	194823952	0.036	0.035	1	90	22	0	0.018	A:C
23	rs6698581	194824387	0	0	1	97.8	28	0	0	A:A
24	rs12409423	194825173	0	0	1	100	30	0	0	C:C
25	rs4633231	194826209	0.125	0.117	1	90	22	0	0.062	G:A
26	rs4348670	194827009	0	0	1	100	30	0	0	T:T
27	rs12131846	194827241	0	0	1	100	30	0	0	A:A
28	rs13375443	194827745	0.036	0.035	1	90	22	0	0.018	T:C
29	rs13375567	194829527	0.034	0.033	1	98.9	29	0	0.017	T:C
30	rs4601531	194829767	0.133	0.124	1	100	30	0	0.067	T:G
31	rs16840219	194829831	0	0	1	100	30	0	0	T:T
32	rs7514111	194831048	0	0	1	98.9	29	0	0	C:C
33	rs12136798	194831444	0	0	1	97.8	28	0	0	T:T
34	rs12117379	194831536	0	0	1	100	30	0	0	C:C
35	rs4484881	194833143	0	0	1	98.9	29	0	0	A:A
36	rs4411090	194833324	0	0	1	98.9	29	0	0	C:C
37	rs12729192	194833436	0	0	1	100	30	0	0	A:A
38	rs12733498	194833486	0	0	1	98.9	29	0	0	T:T
39	rs4614205	194834320	0.153	0.141	1	97.8	28	0	0.076	A:G

FIG. 14

40	rs12144706	194834497	0	0	1	100	30	0	0	C:C
41	rs4658044	194835779	0	0	1	100	30	0	0	G:G
42	rs16840224	194836304	0.339	0.384	0.5423	90	22	0	0.259	C:T
43	rs10754196	194837329	0.357	0.392	0.6745	90	22	0	0.268	A:G
44	rs4658045	194839446	0	0	1	97.8	28	0	0	A:A
45	rs10754198	194840128	0.356	0.403	0.5125	98.9	29	0	0.28	A:T
46	rs1928434	194840617	0	0	1	100	30	0	0	T:T
47	rs13376190	194840780	0	0	1	100	30	0	0	A:A
48	rs12095730	194841198	0	0	1	100	30	0	0	G:G
49	rs6428348	194841546	0	0	1	97.8	28	0	0	A:A
50	rs6428349	194842225	0.429	0.436	1	90	22	0	0.321	A:G
51	rs1360143	194842987	0.15	0.139	1	100	30	0	0.075	G:C
52	rs16840262	194843934	0	0	1	100	30	0	0	T:T
53	rs6682755	194844871	0	0	1	100	30	0	0	A:A
54	rs2183225	194845339	0.033	0.033	1	100	30	0	0.017	C:A
55	rs10801551	194846662	0.232	0.282	0.3462	90	22	0	0.17	G:A
56	rs12142425	194846997	0	0	1	97.8	28	0	0	T:T
57	rs6701301	194847635	0.036	0.035	1	94.4	26	0	0.018	G:A
58	rs4507960	194848005	0.036	0.035	1	90	22	0	0.018	T:C
59	rs16840281	194849285	0	0	1	98.9	29	0	0	A:A
60	rs1360144	194849398	0.036	0.036	1	88.9	21	0	0.018	A:G
61	rs12095119	194849480	0	0	1	98.9	29	0	0	A:A
62	rs6700164	194850230	0	0	1	100	30	0	0	A:A
63	rs4657825	194850944	0.283	0.289	1	100	30	0	0.175	G:A
64	rs12061508	194851091	0.232	0.282	0.3462	90	22	0	0.17	G:A
65	rs16840286	194851092	0	0	1	96.7	27	0	0	A:A
66	rs12117069	194851576	0	0	1	98.9	29	0	0	T:T
67	rs7412736	194851589	0	0	1	98.9	29	0	0	G:G
68	rs944041	194851840	0.036	0.035	1	90	22	0	0.018	G:A
69	rs1887972	194851986	0	0	1	100	30	0	0	G:G
70	rs6691254	194852001	0	0	1	96.7	28	0	0	C:C
71	rs6676445	194852314	0	0	1	97.8	28	0	0	C:C
72	rs6676634	194852444	0.037	0.036	1	86.7	21	1	0.019	C:T
73	rs6702804	194852674	0	0	1	96.7	27	0	0	G:G
74	rs12142359	194852982	0	0	1	100	30	0	0	A:A
75	rs6428353	194854323	0.034	0.033	1	98.9	29	0	0.017	A:T
76	rs6428354	194854443	0.033	0.033	1	100	30	0	0.017	T:C
77	rs12047153	194854535	0	0	1	100	30	0	0	A:A
78	rs6428355	194854598	0.036	0.035	1	90	22	0	0.018	C:A

FIG. 14 CONT.

79	rs1571491	194855366	0.033	0.033	1	100	30	0	0.017	T:A
80	rs1571490	194855586	0.036	0.035	1	90	22	0	0.018	T:C
81	rs12061098	194856190	0	0	1	98.9	29	0	0	G:G
82	rs7520245	194856412	0	0	1	98.9	29	0	0	G:G
83	rs7523133	194856996	0	0	1	100	30	0	0	G:G
84	rs1329425	194857969	0	0	1	100	30	0	0	T:T
85	rs574244	194859846	0.033	0.033	1	100	30	0	0.017	A:G
86	rs12741430	194860321	0.039	0.038	1	86.7	22	0	0.02	A:T
87	rs471554	194860658	0.018	0.018	1	95.6	26	0	0.009	C:A
88	rs542830	194861022	0.036	0.035	1	90	22	0	0.018	A:G
89	rs12121093	194861408	0.018	0.018	1	88.9	21	0	0.009	A:G
90	rs12121930	194861457	0	0	1	96.7	27	0	0	T:T
91	rs7511758	194861924	0	0	1	100	30	0	0	A:A
92	rs7512032	194862148	0	0	1	100	30	0	0	A:A
93	rs6659401	194862789	0	0	1	93.3	25	0	0	A:A
94	rs6670500	194862829	0	0	1	96.7	27	0	0	G:G
95	rs6696102	194863361	0	0	1	98.9	29	0	0	C:C
96	rs493104	194863473	0.033	0.033	1	100	30	0	0.017	G:T
97	rs10489459	194864171	0	0	1	100	30	0	0	G:G
98	rs2143913	194866985	0	0	1	93.3	25	0	0	A:A
99	rs6673512	194867156	0	0	1	97.8	28	0	0	A:A
100	rs12064775	194867228	0.035	0.034	1	95.6	26	0	0.018	A:G
101	rs6676510	194867480	0	0	1	100	30	0	0	T:T
102	rs522637	194867485	0	0	1	100	30	0	0	A:A
103	rs10489458	194868948	0	0	1	100	30	0	0	T:T
104	rs487681	194869029	0.033	0.033	1	100	30	0	0.017	A:T
105	rs10489457	194869137	0	0	1	100	30	0	0	C:C
106	rs12124091	194869564	0	0	1	100	30	0	0	A:A
107	rs12029571	194869925	0.216	0.278	0.2407	87.8	22	1	0.167	A:G
108	rs6695338	194870540	0	0	1	100	30	0	0	G:G
109	rs512154	194871445	0.034	0.033	1	98.9	29	0	0.017	T:A
110	rs6675046	194872128	0	0	1	100	30	0	0	T:T
111	rs11802272	194873182	0	0	1	97.8	28	0	0	T:T
112	rs12133754	194873556	0	0	1	100	30	0	0	C:C
113	rs12067129	194873669	0	0	1	97.8	28	0	0	A:A
114	rs565088	194874781	0.033	0.033	1	100	30	0	0.017	A:G
115	rs17573790	194876019	0.2	0.206	1	100	30	0	0.117	A:C
116	rs16840355	194876583	0	0	1	100	30	0	0	A:A
117	rs479924	194877103	0.033	0.033	1	100	30	0	0.017	T:C

FIG. 14 CONT.

118	rs6693167	194878153	0	0	1	95.6	26	0	0	A:A
119	rs524312	194878370	0.037	0.036	1	93.3	24	0	0.019	T:C
120	rs480773	194878734	0.033	0.033	1	100	30	0	0.017	A:G
121	rs6659034	194879236	0	0	1	97.8	28	0	0	G:G
122	rs2336220	194882318	0	0	1	100	30	0	0	A:A
123	rs512822	194882461	0.036	0.035	1	90	22	0	0.018	G:T
124	rs479718	194883727	0	0	1	98.9	29	0	0	G:G
125	rs505102	194886125	0.255	0.298	0.4653	88.9	21	0	0.182	T:C
126	rs3753394	194887540	0.357	0.423	0.3557	90	22	0	0.304	C:T
127	rs7526622	194888515	0	0	1	100	30	0	0	G:G
128	rs512900	194888987	0.033	0.033	1	100	30	0	0.017	T:C
129	rs2143912	194889247	0	0	1	98.9	29	0	0	C:C
130	rs487114	194889524	0.034	0.033	1	98.9	29	0	0.017	A:C
131	rs7524776	194889960	0.214	0.219	1	90	22	0	0.125	T:C
132	rs6428356	194890583	0	0	1	100	30	0	0	G:G
133	rs508505	194892368	0	0	1	100	30	0	0	G:G
134	rs12134975	194892620	0	0	1	90	23	0	0	G:G
135	rs12746361	194893426	0	0	1	93.3	26	0	0	G:G
136	rs7551203	194893726	0.033	0.033	1	98.9	29	0	0.017	C:T
137	rs16840394	194894818	0	0	1	90	22	0	0	T:T
138	rs12238983	194895889	0	0	1	100	30	0	0	C:C
139	rs521605	194896059	0	0	1	91.1	23	0	0	A:A
140	rs499807	194896127	0.033	0.033	1	100	30	0	0.017	A:G
141	rs6680396	194899093	0.232	0.282	0.3462	90	22	0	0.17	A:G
142	rs16840397	194899328	0	0	1	97.8	29	0	0	G:G
143	rs564657	194900229	0.017	0.017	1	100	30	0	0.008	T:A
144	rs6689826	194901166	0	0	1	97.8	28	0	0	T:T
145	rs529825	194901729	0.267	0.299	0.5935	100	30	0	0.183	G:A
146	rs6691749	194906684	0	0	1	100	30	0	0	T:T
147	rs11579439	194907222	0	0	1	100	30	0	0	C:C
148	rs16840401	194907880	0	0	1	90	22	0	0	A:A
149	rs800292	194908856	0.286	0.316	0.6845	90	22	0	0.196	G:A
150	rs495222	194910006	0	0	1	90	22	0	0	C:C
151	rs1329424	194912799	0.417	0.477	0.44	100	30	0	0.392	G:T
152	rs572515	194912884	0.429	0.469	0.6669	90	22	0	0.375	G:A
153	rs1329423	194913010	0.321	0.423	0.1228	90	22	0	0.304	T:C
154	rs12116702	194915137	0	0	1	97.8	28	0	0	A:A
155	rs12127759	194915236	0.183	0.267	0.0575	100	30	0	0.158	C:T
156	rs766001	194917092	0	0	1	97.8	28	0	0	T:T

FIG. 14 CONT.

157	rs16840419	194918368	0.169	0.259	0.0383	98.9	29	0	0.153	A:G
158	rs3766404	194918455	0.167	0.239	0.0979	86.7	21	1	0.139	T:C
159	rs16840422	194919457	0.183	0.267	0.0575	100	30	0	0.158	C:T
160	rs1061147	194920947	0.429	0.469	0.6669	90	22	0	0.375	C:A
161	rs490864	194921465	0	0	1	100	30	0	0	T:T
162	rs1329422	194921903	0.421	0.474	0.5246	96.7	27	0	0.386	G:C
163	rs2300430	194922336	0.433	0.473	0.6571	98.9	29	0	0.383	C:T
164	rs10801553	194922366	0.429	0.469	0.6669	90	22	0	0.375	C:A
165	rs1329421	194922828	0.436	0.48	0.6412	94.4	25	0	0.4	T:A
166	rs544889	194923205	0	0	1	94.4	25	0	0	A:A
167	rs10801554	194924278	0.434	0.454	0.9164	91.1	23	0	0.349	T:C
168	rs7529589	194924902	0.429	0.469	0.6669	90	22	0	0.375	C:T
169	rs482934	194925120	0.429	0.469	0.6669	90	22	0	0.375	T:G
170	rs1061170	194925860	0.564	0.405	0.005	93.3	25	0	0.282	T:C
171	rs10801555	194926884	0.429	0.469	0.6669	90	22	0	0.375	G:A
172	rs10801556	194927087	0.429	0.469	0.6669	90	22	0	0.375	G:A
173	rs10922094	194928128	0.417	0.477	0.44	100	30	0	0.392	C:G
174	rs12124794	194928161	0.22	0.27	0.3051	98.9	29	0	0.161	A:T
175	rs12405238	194928236	0.339	0.416	0.2598	90	22	0	0.295	G:T
176	rs10922096	194929082	0.429	0.497	0.4018	90	22	0	0.464	T:C
177	rs12041668	194929670	0.196	0.237	0.4346	88.9	20	0	0.137	C:T
178	rs514943	194930536	0.429	0.469	0.6669	90	22	0	0.375	T:C
179	rs579745	194931199	0	0	1	98.9	29	0	0	C:C
180	rs10922102	194934910	0.397	0.475	0.2932	97.8	28	0	0.388	T:C
181	rs2860102	194934942	0.417	0.477	0.44	100	30	0	0.392	T:A
182	rs4658046	194937380	0.429	0.469	0.6669	90	22	0	0.375	T:C
183	rs10754199	194937462	0.429	0.469	0.6669	90	22	0	0.375	G:A
184	rs12565418	194938532	0	0	1	100	30	0	0	T:T
185	rs12038333	194939077	0.429	0.469	0.6669	90	22	0	0.375	A:G
186	rs12045503	194939096	0.414	0.471	0.4738	97.8	28	0	0.379	T:C
187	rs9970784	194940425	0.424	0.495	0.3628	98.9	29	0	0.449	T:C
188	rs1831282	194940616	0.446	0.492	0.621	90	22	0	0.438	C:A
189	rs203687	194940893	0.446	0.492	0.621	90	22	0	0.438	T:C
190	rs2019727	194941337	0.327	0.38	0.4511	88.9	21	0	0.255	T:A
191	rs2019724	194941540	0.446	0.492	0.621	90	22	0	0.438	C:T
192	rs1887973	194941802	0.433	0.495	0.4398	100	30	0	0.45	C:G
193	rs6428357	194942194	0.446	0.492	0.621	90	22	0	0.438	G:A
194	rs7513157	194942303	0.089	0.085	1	90	22	0	0.045	G:A
195	rs6695321	194942484	0.357	0.436	0.2613	90	22	0	0.321	A:G

FIG. 14 CONT.

196	rs10733086	194943558	0.446	0.492	0.621	90	22	0	0.438	T:A
197	rs1410997	194943786	0.446	0.492	0.621	90	22	0	0.438	A:C
198	rs203685	194944568	0.433	0.495	0.4398	100	30	0	0.45	A:C
199	rs203684	194944632	0	0	1	97.8	28	0	0	C:C
200	rs10737680	194946078	0.375	0.487	0.1278	90	22	0	0.42	A:C
201	rs5002880	194947344	0	0	1	97.8	28	0	0	T:T
202	rs1831281	194947437	0.233	0.278	0.3806	100	30	0	0.167	C:T
203	rs7535263	194948969	0.4	0.486	0.2393	100	30	0	0.417	G:A
204	rs1061171	194949629	0.017	0.017	1	97.8	28	0	0.009	C:G
205	rs203678	194949767	0	0	1	90	23	0	0	A:A
206	rs203675	194950783	0	0	1	96.7	27	0	0	A:A
207	rs203674	194951248	0.455	0.489	0.746	87.8	21	0	0.427	T:G
208	rs12085209	194952010	0	0	1	98.9	29	0	0	G:G
209	rs16840462	194952036	0	0	1	93.3	24	0	0	G:G
210	rs16840465	194952599	0	0	1	100	30	0	0	A:A
211	rs3753395	194953275	0.383	0.483	0.1601	100	30	0	0.408	A:T
212	rs6677604	194953541	0.321	0.375	0.424	90	22	0	0.25	G:A
213	rs10489456	194954138	0.183	0.243	0.1576	100	30	0	0.142	G:A
214	rs203673	194954525	0	0	1	92.2	23	0	0	C:C
215	rs2104714	194954704	0	0	1	94.4	25	0	0	G:G
216	rs203671	194955935	0	0	1	95.6	26	0	0	C:C
217	rs203670	194956012	0	0	1	97.8	28	0	0	G:G
218	rs203669	194956617	0	0	1	95.6	26	0	0	C:C
219	rs10922106	194958087	0.415	0.491	0.3559	90	23	0	0.434	A:G
220	rs11801630	194958771	0.175	0.241	0.126	94.4	25	0	0.14	C:T
221	rs12047565	194959032	0	0	1	97.8	28	0	0	G:G
222	rs393955	194959093	0.396	0.494	0.2174	90	24	1	0.443	A:C
223	rs381974	194959295	0.404	0.494	0.2327	96.7	27	0	0.447	G:A
224	rs12096637	194959747	0	0	1	96.7	27	0	0	G:G
225	rs379489	194960074	0.455	0.494	0.6963	88.9	22	0	0.445	G:A
226	rs1474792	194960330	0	0	1	97.8	28	0	0	C:C
227	rs3753396	194962365	0.161	0.232	0.0885	90	22	0	0.134	A:G
228	rs7537967	194962701	0	0	1	85.6	20	0	0	T:T
229	rs7535653	194962757	0	0	1	94.4	25	0	0	A:A
230	rs403846	194963360	0.429	0.49	0.4631	95.6	26	0	0.429	C:T
231	rs419137	194963498	0.179	0.219	0.3706	90	22	0	0.125	T:G
232	rs1410996	194963556	0.383	0.483	0.1601	100	30	0	0.408	G:A
233	rs1060821	194964201	0	0	1	96.7	27	0	0	G:G
234	rs395544	194964895	0.446	0.492	0.621	90	22	0	0.438	G:A

FIG. 14 CONT.

235	rs1576340	194965334	0.185	0.252	0.1457	91.1	23	0	0.148	G:T
236	rs12144939	194965568	0.321	0.337	0.9537	95.6	26	0	0.214	G:T
237	rs11799595	194966945	0.216	0.278	0.2407	85.6	19	0	0.167	G:C
238	rs7514261	194967537	0	0	1	98.9	29	0	0	G:G
239	rs380390	194967674	0.433	0.495	0.4398	100	30	0	0.45	C:G
240	rs380060	194967751	0	0	1	86.7	20	0	0	G:G
241	rs7540032	194967907	0.364	0.487	0.094	92.2	24	0	0.418	C:T
242	rs2284664	194969148	0.245	0.294	0.3947	86.7	19	0	0.179	C:T
243	rs1329428	194969433	0.375	0.487	0.1278	90	22	0	0.42	C:T
244	rs7413137	194970261	0	0	1	97.8	28	0	0	G:G
245	rs70620	194971620	0.183	0.243	0.1576	100	30	0	0.142	G:A
246	rs434536	194971790	0	0	1	90	22	0	0	C:C
247	rs742855	194972143	0.179	0.245	0.1321	90	22	0	0.143	T:C
248	rs428060	194972823	0	0	1	98.9	29	0	0	C:C
249	rs515299	194973300	0	0	1	100	30	0	0	G:G
250	rs11799956	194973489	0	0	1	97.8	28	0	0	G:G
251	rs543879	194973638	0.019	0.018	1	87.8	21	0	0.009	G:A
252	rs408497	194974795	0	0	1	97.8	28	0	0	T:T
253	rs11799380	194975078	0.212	0.273	0.229	85.6	19	0	0.163	A:G
254	rs454652	194975263	0	0	1	98.9	29	0	0	G:G
255	rs405306	194975318	0	0	1	98.9	29	0	0	C:C
256	rs424535	194975846	0.429	0.477	0.5804	90	22	0	0.393	A:T
257	rs1065489	194976397	0.182	0.249	0.1387	88.9	22	0	0.145	G:T
258	rs11582939	194976780	0.179	0.245	0.1321	90	22	0	0.143	C:T
259	rs385892	194977413	0	0	1	97.8	28	0	0	T:T
260	rs16840522	194977539	0.321	0.375	0.424	90	22	0	0.25	T:C
261	rs17575212	194977661	0	0	1	95.6	26	0	0	A:A
262	rs534399	194977690	0	0	1	100	30	0	0	G:G
263	rs11539862	194977699	0	0	1	96.7	27	0	0	G:G
264	rs1040597	194977722	0	0	1	98.9	29	0	0	T:T
265	rs513699	194979200	0	0	1	95.6	26	0	0	T:T
266	rs422851	194979562	0	0	1	94.4	25	0	0	C:C
267	rs10801560	194981223	0.233	0.278	0.3806	100	30	0	0.167	C:A
268	rs10801561	194981293	0.233	0.278	0.3806	100	30	0	0.167	T:A
269	rs460184	194982960	0.05	0.049	1	100	30	0	0.025	T:C
270	rs488738	194983244	0	0	1	100	30	0	0	T:T
271	rs386258	194983553	0	0	1	94.4	26	0	0	C:C
272	rs403990	194984668	0	0	1	100	30	0	0	A:A
273	rs402032	194984837	0.35	0.413	0.3437	100	30	0	0.292	C:G

FIG. 14 CONT.

274	rs455497	194989670	0	0	1	100	30	0	0	T:T
275	rs401808	194995218	0	0	1	100	30	0	0	C:C
276	rs449847	194997771	0.228	0.202	0.9257	96.7	27	0	0.114	A:T
277	rs425173	194997796	0	0	1	98.9	29	0	0	A:A
278	rs421581	195002856	0	0	1	91.1	24	0	0	C:C
279	rs12046285	195005393	0	0	1	98.9	29	0	0	C:C
280	rs369816	195005578	0	0	1	100	30	0	0	G:G
281	rs395998	195006460	0.433	0.495	0.4398	100	30	0	0.45	T:C
282	rs385532	195007022	0	0	1	96.7	27	0	0	G:G
283	rs385390	195010550	0.183	0.243	0.1576	100	30	0	0.142	A:C
284	rs17575274	195015067	0	0	1	88.9	23	0	0	C:C
285	rs445207	195016368	0.25	0.33	0.1269	100	30	0	0.208	T:C
286	rs370789	195022863	0.164	0.286	0.0099	92.2	25	0	0.173	A:G
287	rs421820	195022923	0.182	0.222	0.3822	88.9	21	0	0.127	C:T
288	rs378283	195024909	0.167	0.29	0.0109	91.1	24	0	0.176	T:C
289	rs389897	195025053	0.233	0.32	0.0861	100	30	0	0.2	A:T
290	rs377298	195025164	0.176	0.327	0.0054	85.6	20	0	0.206	A:C
291	rs12738227	195026780	0.019	0.019	1	87.8	21	0	0.01	C:A
292	rs643781	195026803	0.173	0.299	0.013	85.6	20	1	0.183	A:C
293	rs426736	195027040	0.169	0.234	0.1147	98.9	29	0	0.136	A:G
294	rs433142	195028383	0	0	1	98.9	29	0	0	G:G
295	rs422427	195028709	0	0	1	90	22	0	0	G:G
296	rs411854	195028740	0.467	0.48	0.988	100	30	0	0.4	A:G
297	rs370953	195030988	0.182	0.222	0.3822	87.8	21	0	0.127	T:C
298	rs9427913	195032090	0.4	0.495	0.1946	100	30	0	0.45	G:T
299	rs644598	195033200	0.2	0.231	0.5261	100	30	0	0.133	T:C
300	rs1969893	195036863	0	0	1	97.8	28	0	0	T:T
301	rs564004	195037161	0.232	0.205	0.9114	88.9	22	0	0.116	T:C
302	rs384006	195037748	0.054	0.052	1	90	22	0	0.027	G:A
303	rs371075	195043459	0.186	0.247	0.1648	97.8	28	0	0.144	T:C
304	rs446713	195048499	0.233	0.206	0.8491	100	30	0	0.117	T:C
305	rs436719	195054367	0.35	0.413	0.3437	100	30	0	0.292	T:G
306	rs7523582	195054565	0	0	1	94.4	26	0	0	A:A
307	rs432007	195059087	0.417	0.499	0.2776	100	30	0	0.475	C:A
308	rs438781	195062863	0.448	0.498	0.5757	96.7	29	1	0.466	A:T
309	rs677949	195063750	0	0	1	93.3	25	0	0	T:T
310	rs388145	195064170	0	0	1	92.2	24	0	0	G:G
311	rs364806	195065395	0	0	1	100	30	0	0	T:T
312	rs419436	195065612	0.054	0.052	1	90	22	0	0.027	C:T

FIG. 14 CONT.

313	rs3179476	195066260	0.296	0.366	0.2723	86.7	20	0	0.241	G:A
314	rs1984954	195066889	0	0	1	98.9	29	0	0	A:A
315	rs380556	195068574	0	0	1	93.3	25	0	0	C:C
316	rs1738740	195075263	0.467	0.495	0.8032	100	30	0	0.45	T:A
317	rs800249	195076539	0	0	1	100	30	0	0	C:C
318	rs464967	195076863	0	0	1	88.9	22	0	0	T:T
319	rs6679884	195084621	0.305	0.259	0.433	98.9	29	0	0.153	A:G
320	rs503002	195086910	0.35	0.413	0.3437	100	30	0	0.292	G:A
321	rs494636	195086917	0	0	1	98.9	29	0	0	A:A
322	rs10754206	195087547	0.431	0.498	0.4522	85.6	20	0	0.471	T:G
323	rs1964645	195088637	0.17	0.243	0.1031	86.7	20	0	0.142	C:T
324	rs1963605	195088791	0.267	0.299	0.5935	100	30	0	0.183	A:C
325	rs16840607	195089653	0.377	0.449	0.3542	86.7	20	0	0.34	T:G
326	rs10922144	195089923	0.25	0.293	0.4466	90	22	0	0.179	C:T
327	rs7542235	195090236	0.321	0.375	0.424	90	22	0	0.25	A:G
328	rs16840616	195090339	0	0	1	86.7	20	0	0	A:A
329	rs16840639	195091396	0.317	0.367	0.4313	100	30	0	0.242	T:C
330	rs16840645	195091700	0	0	1	100	30	0	0	A:A
331	rs7519758	195091910	0.31	0.348	0.5873	95.6	28	0	0.224	C:T
332	rs12562460	195092060	0	0	1	98.9	29	0	0	C:C
333	rs16840658	195092251	0.125	0.205	0.034	90	22	0	0.116	G:A
334	rs16840661	195092486	0	0	1	90	22	0	0	T:T
335	rs16840666	195093011	0	0	1	100	30	0	0	A:A
336	rs12728643	195093856	0	0	1	100	30	0	0	A:A
337	rs17494275	195095494	0.291	0.341	0.4282	87.8	21	0	0.218	T:A
338	rs17269692	195095522	0	0	1	100	30	0	0	G:G
339	rs16840669	195095561	0	0	1	87.8	21	0	0	G:G
340	rs6682736	195097555	0	0	1	97.8	28	0	0	C:C
341	rs7412359	195099548	0.186	0.247	0.1648	97.8	28	0	0.144	G:A
342	rs10922146	195101259	0.25	0.289	0.4814	100	30	0	0.175	C:T
343	rs6428361	195101703	0	0	1	100	30	0	0	C:C
344	rs12047098	195101729	0.255	0.298	0.4653	87.8	21	0	0.182	T:C
345	rs6681234	195102101	0	0	1	100	30	0	0	C:C
346	rs6656059	195102121	0	0	1	100	30	0	0	G:G
347	rs7552547	195103622	0.2	0.255	0.2185	100	30	0	0.15	T:G
348	rs6657442	195104683	0.25	0.293	0.4466	90	22	0	0.179	T:C
349	rs7413265	195105786	0.255	0.298	0.4653	87.8	21	0	0.182	G:T
350	rs7516560	195106053	0.182	0.249	0.1387	87.8	21	0	0.145	T:C
351	rs7514704	195106614	0.183	0.243	0.1576	100	30	0	0.142	A:G

FIG. 14 CONT.

352	rs12401879	195106682	0	0	1	98.9	29	0	0	A:A
353	rs7519492	195106748	0.183	0.243	0.1576	100	30	0	0.142	T:C
354	rs7517126	195106895	0.179	0.245	0.1321	90	22	0	0.143	A:G
355	rs7517587	195107388	0.186	0.247	0.1648	98.9	29	0	0.144	A:G
356	rs2016046	195107669	0.017	0.017	1	100	30	0	0.008	T:C
357	rs10737682	195108253	0	0	1	100	30	0	0	A:A
358	rs13375144	195108313	0.179	0.245	0.1321	90	22	0	0.143	C:T
359	rs13375150	195108462	0.186	0.247	0.1648	98.9	29	0	0.144	C:T
360	rs13374399	195108629	0.183	0.243	0.1576	100	30	0	0.142	G:A
361	rs13375236	195108894	0.182	0.249	0.1387	87.8	21	0	0.145	C:T
362	rs2336502	195109197	0.383	0.46	0.2807	100	30	0	0.358	T:C
363	rs2133138	195109794	0.182	0.249	0.1387	87.8	21	0	0.145	G:A
364	rs2336503	195110043	0.179	0.245	0.1321	90	22	0	0.143	C:A
365	rs2336505	195110278	0	0	1	95.6	27	0	0	A:A
366	rs6428363	195110334	0.364	0.463	0.1705	87.8	21	0	0.364	G:A
367	rs6660329	195110358	0	0	1	100	30	0	0	T:T
368	rs1542839	195110458	0	0	1	100	30	0	0	C:C
369	rs6428364	195110672	0.182	0.249	0.1387	87.8	21	0	0.145	C:T
370	rs6428365	195110754	0.19	0.225	0.4465	95.6	28	1	0.129	C:A
371	rs6428366	195110790	0.182	0.249	0.1387	87.8	21	0	0.145	A:G
372	rs6428367	195110815	0.182	0.249	0.1387	87.8	21	0	0.145	T:C
373	rs6428368	195110933	0.186	0.247	0.1648	96.7	27	0	0.144	T:C
374	rs6428369	195110958	0.182	0.249	0.1387	87.8	21	0	0.145	T:C
375	rs6428370	195111216	0.364	0.463	0.1705	87.8	21	0	0.364	A:G
376	rs12240143	195111640	0.375	0.464	0.2218	90	22	0	0.366	T:C
377	rs1021636	195111896	0.264	0.306	0.5058	84.4	20	1	0.189	A:G
378	rs12240148	195111930	0.183	0.243	0.1576	98.9	29	0	0.142	T:C
379	rs6695525	195112144	0.481	0.495	0.9971	80	18	0	0.452	T:G
380	rs11811456	195114034	0.255	0.341	0.1247	87.8	21	0	0.218	A:G
381	rs1995148	195114092	0	0	1	98.9	29	0	0	G:G
382	rs10801575	195119404	0.321	0.459	0.043	90	22	0	0.357	C:T
383	rs7517836	195121926	0	0	1	97.8	28	0	0	C:C
384	rs6428372	195125871	0.283	0.33	0.427	100	30	0	0.208	A:C
385	rs7366238	195127420	0.217	0.315	0.096	82.2	20	1	0.196	A:G
386	rs12031992	195127748	0	0	1	100	30	0	0	G:G
387	rs7416180	195128924	0.02	0.02	1	85.6	21	1	0.01	T:G
388	rs12733105	195128941	0	0	1	98.9	29	0	0	T:T
389	rs12404243	195129192	0.268	0.347	0.1684	88.9	21	0	0.223	A:C
390	rs12734260	195132040	0.211	0.188	1	95.6	27	0	0.105	G:T

FIG. 14 CONT.

391	rs6664223	195132208	0	0	1	98.9	29	0	0	T:T
392	rs11585500	195132728	0.069	0.067	1	96.7	27	0	0.034	C:T
393	rs7522691	195133425	0.017	0.017	1	100	30	0	0.008	C:T
394	rs6685931	195133856	0.429	0.469	0.6669	88.9	21	0	0.375	T:C
395	rs7546940	195137415	0.25	0.349	0.0648	100	30	0	0.225	G:A
396	rs7416336	195138798	0.25	0.349	0.0648	100	30	0	0.225	C:G
397	rs12406509	195140815	0.071	0.069	1	90	22	0	0.036	A:G
398	rs7417769	195143081	0.345	0.468	0.085	93.3	24	0	0.373	G:A
399	rs10801579	195143742	0.273	0.37	0.1009	93.3	25	0	0.245	G:A
400	rs10922147	195144312	0.25	0.31	0.2493	100	30	0	0.192	C:G
401	rs1409153	195146628	0.418	0.468	0.5685	88.9	21	0	0.373	T:C
402	rs4998887	195147383	0	0	1	100	30	0	0	C:C
403	rs4044578	195148043	0.407	0.466	0.4758	86.7	20	0	0.37	A:C
404	rs1853883	195148223	0.418	0.489	0.3808	87.8	21	0	0.427	C:G
405	rs7522952	195148967	0.196	0.205	1	90	22	0	0.116	A:G
406	rs7525447	195149059	0.183	0.243	0.1576	98.9	29	0	0.142	T:C
407	rs7533270	195149126	0.19	0.25	0.1724	96.7	27	0	0.147	G:A
408	rs1325927	195150304	0	0	1	98.9	29	0	0	T:T
409	rs1853882	195151967	0.183	0.243	0.1576	100	30	0	0.142	C:G
410	rs10801580	195152308	0.255	0.32	0.242	87.8	21	0	0.2	T:C
411	rs2477997	195153066	0	0	1	92.2	24	0	0	T:T
412	rs4915559	195153393	0.255	0.32	0.242	87.8	21	0	0.2	T:C
413	rs1971579	195153804	0.441	0.424	1	97.8	28	0	0.305	T:G
414	rs3795341	195153897	0.305	0.324	0.8693	97.8	28	0	0.203	G:A
415	rs12758862	195154007	0	0	1	97.8	28	0	0	C:C
416	rs12744282	195154029	0	0	1	97.8	28	0	0	T:T
417	rs10494745	195154080	0.125	0.148	0.5881	90	22	0	0.08	G:A
418	rs12757822	195154100	0	0	1	96.7	27	0	0	G:G
419	rs9427640	195154385	0	0	1	97.8	28	0	0	G:G
420	rs7548070	195158945	0.179	0.245	0.1321	90	22	0	0.143	G:A
421	rs4086175	195160949	0.196	0.232	0.4732	90	22	0	0.134	T:C
422	rs3906115	195161157	0.364	0.472	0.1358	92.2	23	0	0.382	A:T
423	rs4915318	195163711	0.25	0.316	0.2283	90	22	0	0.196	C:A
424	rs7538501	195168376	0.179	0.245	0.1321	90	22	0	0.143	C:T
425	rs2986127	195171294	0.317	0.367	0.4313	100	30	0	0.242	C:T
426	rs388338	195171556	0.241	0.366	0.0247	96.7	29	1	0.241	C:A
427	rs432366	195178814	0.35	0.413	0.3437	100	30	0	0.292	C:G
428	rs9427929	195178866	0.2	0.206	1	100	30	0	0.117	C:G
429	rs1853881	195182368	0.193	0.254	0.1804	93.3	26	0	0.149	A:C

FIG. 14 CONT.

430	rs12753081	195182867	0	0	1	100	30	0	0	C:C
431	rs12752032	195182942	0	0	1	98.9	29	0	0	G:G
432	rs6674522	195182982	0.196	0.205	1	90	22	0	0.116	G:C
433	rs9427649	195183076	0	0	1	100	30	0	0	G:G
434	rs10494746	195183509	0.067	0.064	1	100	30	0	0.033	T:A
435	rs12066959	195184522	0.288	0.322	0.6552	87.8	21	0	0.202	G:A
436	rs12746391	195185623	0	0	1	95.6	26	0	0	A:A
437	rs4085749	195186771	0.271	0.303	0.6148	98.9	29	0	0.186	C:T
438	rs3828032	195186801	0.321	0.459	0.043	90	22	0	0.357	C:T
439	rs3790414	195186922	0.267	0.299	0.5935	100	30	0	0.183	T:A
440	rs3790413	195187161	0	0	1	98.9	29	0	0	A:A
441	rs9427933	195188981	0	0	1	100	30	0	0	C:C
442	rs9427934	195189483	0.333	0.455	0.0639	100	30	0	0.35	G:A
443	rs12027398	195191920	0	0	1	100	30	0	0	A:A
444	rs12072040	195192780	0	0	1	100	30	0	0	G:G
445	rs3748556	195193579	0.321	0.459	0.043	90	22	0	0.357	C:T
446	rs3748555	195193596	0	0	1	100	30	0	0	C:C
447	rs7531555	195195933	0.314	0.338	0.8225	84.4	19	0	0.216	C:T
448	rs12723250	195196717	0.321	0.459	0.043	90	22	0	0.357	C:T
449	rs9427653	195198942	0	0	1	100	30	0	0	A:A
450	rs9427938	195199868	0.196	0.205	1	90	22	0	0.116	C:T
451	rs4915546	195200710	0	0	1	100	30	0	0	C:C
452	rs7548145	195202319	0.185	0.198	0.9924	91.1	23	0	0.111	T:C
453	rs7417200	195202448	0	0	1	100	30	0	0	A:A
454	rs9427939	195203105	0	0	1	100	30	0	0	G:G
455	rs6428379	195204159	0.446	0.496	0.5834	90	22	0	0.455	C:T
456	rs6687080	195204235	0	0	1	100	30	0	0	C:C
457	rs6687192	195204303	0	0	1	100	30	0	0	C:C
458	rs10737684	195205813	0.068	0.065	1	97.8	28	0	0.034	A:G
459	rs6669207	195206465	0.317	0.349	0.6576	100	30	0	0.225	G:A
460	rs6663662	195207025	0	0	1	100	30	0	0	T:T
461	rs6667243	195208116	0.464	0.499	0.744	90	22	0	0.482	T:C
462	rs6675769	195208284	0.446	0.5	0.5473	90	22	0	0.491	G:A
463	rs13376702	195209244	0.179	0.191	0.9644	88.9	21	0	0.107	C:T
464	rs9427656	195209643	0.183	0.193	1	100	30	0	0.108	A:T
465	rs9427657	195209944	0.179	0.191	0.9644	90	22	0	0.107	A:G
466	rs9427940	195209976	0.186	0.169	1	98.9	29	0	0.093	G:T
467	rs7539363	195210962	0	0	1	100	30	0	0	G:G
468	rs10801582	195210980	0.271	0.282	1	97.8	28	0	0.169	G:A

FIG. 14 CONT.

469	rs6702340	195211673	0.15	0.139	1	100	30	0	0.075	G:T
470	rs6702632	195211943	0	0	1	100	30	0	0	G:G
471	rs6694672	195212412	0.161	0.148	1	90	22	0	0.08	T:G
472	rs6692162	195212535	0.15	0.139	1	100	30	0	0.075	A:T
473	rs9427660	195213043	0.153	0.141	1	98.9	29	0	0.076	T:A
474	rs9427942	195213071	0.161	0.148	1	90	22	0	0.08	G:A
475	rs9427661	195213169	0.161	0.148	1	90	22	0	0.08	T:C
476	rs9427662	195213398	0.161	0.148	1	90	22	0	0.08	T:C
477	rs3748557	195213492	0.317	0.349	0.6576	100	30	0	0.225	T:A
478	rs12755054	195213653	0.411	0.43	0.9183	90	22	0	0.312	T:C
479	rs12731209	195213762	0.153	0.141	1	98.9	29	0	0.076	C:T
480	rs1170880	195213937	0.161	0.148	1	90	22	0	0.08	T:C
481	rs11583363	195215070	0.05	0.049	1	100	30	0	0.025	A:C
482	rs1170881	195215894	0.15	0.139	1	98.9	29	0	0.075	G:A
483	rs928440	195216973	0.15	0.139	1	100	30	0	0.075	G:A
484	rs928439	195217105	0.153	0.141	1	98.9	29	0	0.076	G:A
485	rs12097550	195218715	0.017	0.017	1	97.8	28	0	0.009	C:T
486	rs7555407	195219029	0	0	1	100	30	0	0	C:C
487	rs1759016	195219121	0.411	0.43	0.9183	90	22	0	0.312	C:T
488	rs7532441	195219192	0.017	0.017	1	100	30	0	0.008	G:A
489	rs1750311	195220848	0.411	0.43	0.9183	90	22	0	0.312	C:A
490	rs16840943	195228576	0	0	1	100	30	0	0	G:G
491	rs7537588	195229605	0.067	0.064	1	100	30	0	0.033	G:A
492	rs10922152	195229629	0.448	0.49	0.6478	96.7	28	0	0.431	A:T
493	rs7528757	195230218	0	0	1	93.3	24	0	0	C:C
494	rs7550650	195230417	0	0	1	96.7	27	0	0	G:G
495	rs7550735	195230471	0	0	1	98.9	29	0	0	G:G
496	rs9727516	195232728	0.017	0.017	1	97.8	28	0	0.008	A:T
497	rs12092294	195233476	0.161	0.148	1	90	22	0	0.08	A:G
498	rs12745733	195236800	0	0	1	98.9	29	0	0	T:T
499	rs12097879	195237056	0	0	1	98.9	29	0	0	G:G
500	rs7554757	195239677	0.109	0.103	1	93.3	24	0	0.055	T:A
501	rs12116643	195239806	0.214	0.245	0.5869	90	22	0	0.143	T:C
502	rs2151136	195241687	0	0	1	100	30	0	0	G:G
503	rs16840956	195244312	0	0	1	97.8	28	0	0	T:T
504	rs10922153	195245238	0.527	0.499	0.9387	88.9	22	0	0.482	G:T
505	rs6663083	195247283	0.491	0.497	1	88.9	22	0	0.464	C:T
506	rs7555070	195248777	0.411	0.43	0.9183	90	22	0	0.312	T:C
507	rs1332665	195248915	0	0	1	100	30	0	0	G:G

FIG. 14 CONT.

508	rs6670056	195249399	0.15	0.139	1	100	30	0	0.075	C:T
509	rs12745275	195249888	0	0	1	100	30	0	0	A:A
510	rs1332666	195251302	0.482	0.496	0.9936	90	22	0	0.455	G:A
511	rs12403534	195254486	0	0	1	100	30	0	0	G:G
512	rs12028161	195256841	0	0	1	100	30	0	0	T:T
513	rs1576879	195258941	0.15	0.139	1	100	30	0	0.075	G:C
514	rs1576880	195259134	0.153	0.141	1	98.9	29	0	0.076	T:C
515	rs12125782	195259742	0.121	0.113	1	95.6	26	0	0.06	A:T
516	rs2336597	195259962	0.161	0.148	1	90	22	0	0.08	G:A
517	rs1360558	195260596	0.482	0.487	1	90	22	0	0.42	G:A
518	rs2336596	195261715	0.5	0.5	1	90	22	0	0.5	A:A
519	rs1332667	195262471	0.522	0.496	1	83.3	20	0	0.457	T:G
520	rs7545831	195263661	0	0	1	100	30	0	0	G:G
521	rs7538192	195263738	0	0	1	100	30	0	0	T:T
522	rs12039586	195264075	0.15	0.139	1	100	30	0	0.075	G:C
523	rs7418993	195266670	0	0	1	98.9	29	0	0	G:G
524	rs12077334	195266876	0	0	1	92.2	25	0	0	G:G
525	rs2336595	195267099	0.15	0.139	1	100	30	0	0.075	C:T
526	rs7545829	195267406	0	0	1	98.9	29	0	0	A:A
527	rs12075161	195267722	0.232	0.257	0.7093	94.4	27	1	0.152	T:A
528	rs12132141	195268252	0	0	1	100	30	0	0	G:G
529	rs6661664	195268858	0	0	1	100	30	0	0	G:G
530	rs12140782	195269150	0.483	0.46	0.9667	98.9	29	0	0.358	C:G
531	rs7554881	195269890	0	0	1	97.8	28	0	0	A:A
532	rs1412639	195270216	0.153	0.141	1	97.8	28	0	0.076	A:G
533	rs10737685	195271107	0.491	0.5	1	94.4	25	0	0.491	A:G
534	rs6693887	195271201	0	0	1	98.9	29	0	0	C:C
535	rs6691224	195271790	0	0	1	97.8	28	0	0	A:A
536	rs6702252	195271835	0.492	0.5	1	97.8	28	0	0.5	A:A
537	rs1332668	195272359	0.5	0.5	1	90	22	0	0.5	C:C
538	rs1332669	195272506	0.161	0.148	1	90	22	0	0.08	T:C
539	rs12724663	195272875	0	0	1	95.6	26	0	0	G:G
540	rs12724676	195272893	0	0	1	96.7	27	0	0	G:G
541	rs12724830	195272975	0	0	1	98.9	29	0	0	G:G
542	rs13375369	195273852	0	0	1	88.9	22	0	0	G:G
543	rs857021	195274102	0.161	0.148	1	90	22	0	0.08	G:A
544	rs698859	195274988	0.482	0.487	1	90	22	0	0.42	C:T
545	rs5990	195275220	0.5	0.5	1	100	30	0	0.5	A:A
546	rs12134960	195276131	0.217	0.243	0.6395	100	30	0	0.142	G:C

FIG. 14 CONT.

547	rs5998	195276421	0.5	0.5	1	90	22	0	0.5	A:A
548	rs10484502	195277098	0.5	0.5	1	100	30	0	0.5	A:A
549	rs857025	195277418	0.483	0.489	1	100	30	0	0.425	C:T
550	rs10754209	195278200	0.5	0.5	1	90	22	0	0.5	A:A
551	rs12753471	195278219	0	0	1	100	30	0	0	C:C
552	rs7518773	195278259	0.5	0.5	1	90	22	0	0.5	A:A
553	rs12734951	195278277	0	0	1	100	30	0	0	A:A
554	rs10754210	195278734	0.5	0.469	0.8881	90	22	0	0.375	G:A
555	rs857027	195280087	0.017	0.017	1	96.7	27	0	0.008	C:T
556	rs857019	195284454	0	0	1	98.9	29	0	0	A:A
557	rs6000	195286481	0	0	1	100	30	0	0	A:A
558	rs6001	195286560	0	0	1	100	30	0	0	T:T
559	rs2990510	195287281	0.5	0.469	0.8881	90	22	0	0.375	T:G
560	rs10801586	195287812	0.186	0.222	0.434	96.7	27	0	0.127	C:T
561	rs1611982	195289391	0.478	0.5	0.9404	81.1	20	0	0.5	A:A
562	rs1412635	195289671	0.508	0.499	1	98.9	29	0	0.475	G:A
563	rs1412634	195289779	0.136	0.126	1	97.8	28	0	0.068	C:T
564	rs1759010	195290852	0	0	1	100	30	0	0	G:G
565	rs5991	195292774	0	0	1	100	30	0	0	T:T
566	rs5999	195292888	0	0	1	100	30	0	0	T:T
567	rs17514281	195292912	0	0.035	0.018	90	22	0	0.018	A:G
568	rs2990511	195293062	0	0	1	98.9	29	0	0	C:C
569	rs1759009	195293295	0.161	0.148	1	90	22	0	0.08	G:A
570	rs1759008	195293518	0.161	0.148	1	90	22	0	0.08	C:A
571	rs1759007	195294229	0.161	0.148	1	90	22	0	0.08	G:A
572	rs11810678	195294493	0	0	1	100	30	0	0	G:G
573	rs2298883	195295257	0.186	0.222	0.434	97.8	28	0	0.127	A:G
574	rs5994	195296159	0	0	1	100	30	0	0	G:G
575	rs17514253	195296710	0.036	0.035	1	90	22	0	0.018	C:T
576	rs5997	195296824	0.161	0.148	1	88.9	21	0	0.08	C:T
577	rs1615413	195296940	0.5	0.5	1	100	30	0	0.5	A:A
578	rs6003	195297644	0.145	0.135	1	88.9	22	0	0.073	T:C
579	rs1759006	195298287	0.15	0.139	1	100	30	0	0.075	C:T
580	rs12752075	195298674	0	0	1	97.8	28	0	0	A:A
581	rs12732168	195298675	0	0	1	96.7	28	0	0	C:C
582	rs12752091	195298712	0	0	1	97.8	28	0	0	A:A
583	rs6002	195298730	0	0	1	85.6	19	0	0	T:T
584	rs5995	195298842	0	0	1	98.9	29	0	0	A:A
585	rs5996	195298968	0.475	0.488	0.9855	98.9	29	0	0.424	C:G

FIG. 14 CONT.

586	rs1627765	195299666	0.179	0.163	1	90	22	0	0.089	A:G
587	rs2298882	195301449	0.474	0.5	0.8416	95.6	27	0	0.5	G:G
588	rs4915148	195302161	0.161	0.148	1	90	22	0	0.08	C:T
589	rs1412632	195303491	0.161	0.148	1	90	22	0	0.08	A:G
590	rs1412631	195303860	0.161	0.148	1	90	22	0	0.08	C:T
591	rs1115247	195304290	0.15	0.139	1	98.9	29	0	0.075	G:A
592	rs7530627	195311103	0	0	1	100	30	0	0	C:C
593	rs1855115	195313405	0.071	0.069	1	90	22	0	0.036	A:C
594	rs4915309	195315404	0.153	0.141	1	98.9	29	0	0.076	T:A
595	rs4915313	195315978	0.15	0.139	1	100	30	0	0.075	T:G
596	rs1537319	195319427	0.133	0.124	1	100	30	0	0.067	C:T
597	rs1332660	195319764	0.182	0.249	0.1387	88.9	22	0	0.145	T:C
598	rs12677	195319996	0.161	0.148	1	90	22	0	0.08	A:G
599	rs1537318	195320017	0	0	1	100	30	0	0	G:G
600	rs12122415	195321230	0	0	1	100	30	0	0	T:T
601	rs10733087	195322405	0.133	0.124	1	100	30	0	0.067	C:T
602	rs10754213	195322548	0.143	0.133	1	90	22	0	0.071	C:T
603	rs2151133	195324659	0.133	0.124	1	100	30	0	0.067	C:T
604	rs10801587	195325003	0.138	0.128	1	97.8	28	0	0.069	T:C
605	rs6670993	195325031	0	0	1	100	30	0	0	T:T
606	rs12406310	195325555	0	0	1	100	30	0	0	C:C
607	rs7528827	195326005	0	0	1	100	30	0	0	T:T
608	rs4915315	195326501	0.15	0.167	0.7906	100	30	0	0.092	A:G
609	rs10754214	195326515	0.143	0.133	1	90	22	0	0.071	G:A
610	rs4915316	195326551	0.143	0.133	1	90	22	0	0.071	G:A
611	rs955927	195327406	0.5	0.486	1	100	30	0	0.417	A:T
612	rs6428387	195328180	0.133	0.124	1	100	30	0	0.067	G:A
613	rs4915156	195329443	0.133	0.124	1	100	30	0	0.067	T:A
614	rs10801588	195329546	0.105	0.1	1	96.7	27	0	0.053	C:T
615	rs7539642	195330464	0.133	0.124	1	100	30	0	0.067	G:A
616	rs6700180	195331119	0.482	0.5	0.948	90	22	0	0.491	A:G
617	rs1953064	195331449	0.133	0.124	1	100	30	0	0.067	G:A
618	rs4442391	195331487	0	0	1	100	30	0	0	C:C
619	rs3891964	195332697	0.143	0.133	1	90	22	0	0.071	C:T
620	rs10737686	195334823	0.183	0.219	0.422	100	30	0	0.125	C:T
621	rs3762271	195337065	0.482	0.487	1	90	22	0	0.42	G:T
622	rs12138336	195337144	0.121	0.143	0.5696	97.8	28	0	0.078	C:G
623	rs10922162	195337399	0.182	0.222	0.3822	88.9	22	0	0.127	C:T
624	rs1412640	195337438	0.161	0.148	1	90	22	0	0.08	C:T

FIG. 14 CONT.

625	rs964201	195337524	0	0	1	90	22	0	0	G:G
626	rs2878749	195340555	0.482	0.487	1	90	22	0	0.42	T:C
627	rs10801589	195342817	0.5	0.5	1	90	22	0	0.5	C:C
628	rs10922163	195344014	0.508	0.5	1	98.9	29	0	0.492	C:T
629	rs12034362	195344139	0.179	0.219	0.3706	90	22	0	0.125	T:C
630	rs11588283	195344440	0	0	1	97.8	28	0	0	C:C
631	rs6703400	195345023	0.161	0.148	1	90	22	0	0.08	C:T
632	rs4915327	195345753	0.15	0.139	1	100	30	0	0.075	C:T
633	rs2026429	195346294	0.161	0.148	1	90	22	0	0.08	A:C
634	rs12723094	195346609	0	0	1	98.9	29	0	0	A:A
635	rs10922166	195346659	0.15	0.139	1	100	30	0	0.075	C:G
636	rs10801590	195346826	0.153	0.141	1	97.8	28	0	0.076	C:A
637	rs12085377	195348305	0.19	0.225	0.4465	96.7	27	0	0.129	G:A
638	rs6671696	195349819	0.127	0.119	1	92.2	24	0	0.064	C:T
639	rs6680497	195349952	0.15	0.139	1	100	30	0	0.075	A:G
640	rs7411719	195352299	0.15	0.139	1	98.9	29	0	0.075	A:T
641	rs1412641	195352433	0	0	1	100	30	0	0	G:G
642	rs10922168	195352753	0.179	0.219	0.3706	90	22	0	0.125	T:C
643	rs11806768	195352892	0	0	1	100	30	0	0	T:T
644	rs10754215	195353292	0.161	0.148	1	90	22	0	0.08	T:C
645	rs12116571	195355616	0.2	0.206	1	100	30	0	0.117	C:T
646	rs12043195	195355748	0	0	1	100	30	0	0	G:G
647	rs4915337	195358160	0.136	0.126	1	97.8	28	0	0.068	T:A
648	rs16841081	195359984	0	0	1	97.8	28	0	0	G:G
649	rs6676084	195360653	0.5	0.469	0.8881	90	22	0	0.375	C:T
650	rs12042442	195362758	0.19	0.225	0.4465	97.8	28	0	0.129	T:C
651	rs7520177	195363052	0	0	1	97.8	28	0	0	T:T
652	rs7534353	195363103	0.155	0.143	1	97.8	28	0	0.078	G:T
653	rs7534439	195363184	0	0	1	98.9	29	0	0	T:T
654	rs7520405	195363325	0	0	1	100	30	0	0	C:C
655	rs7542397	195363343	0.15	0.139	1	100	30	0	0.075	A:G
656	rs12062590	195363987	0	0	1	100	30	0	0	C:C
657	rs7541125	195365559	0	0	1	100	30	0	0	T:T
658	rs7538717	195365632	0	0	1	100	30	0	0	A:A
659	rs7551108	195365691	0.017	0.017	1	100	30	0	0.008	G:C
660	rs6679189	195367179	0.15	0.139	1	98.9	29	0	0.075	A:C
661	rs10922169	195367728	0.15	0.139	1	100	30	0	0.075	C:T
662	rs3737110	195367926	0.483	0.489	1	100	30	0	0.425	C:A
663	rs4915344	195369368	0.179	0.219	0.3706	90	22	0	0.125	A:G

FIG. 14 CONT.

664	rs7553639	195369650	0	0	1	98.9	29	0	0	A:A
665	rs877897	195370598	0.15	0.139	1	100	30	0	0.075	A:G
666	rs2151134	195372159	0.161	0.148	1	93.3	26	1	0.08	G:T
667	rs16841135	195373820	0	0	1	100	30	0	0	A:A
668	rs6688214	195375020	0	0	1	100	30	0	0	C:C
669	rs1332662	195375050	0.15	0.139	1	100	30	0	0.075	G:A
670	rs17550662	195375569	0.055	0.053	1	92.2	24	0	0.027	A:G
671	rs1332663	195375665	0.161	0.148	1	90	22	0	0.08	T:C
672	rs1888991	195377445	0.179	0.163	1	90	22	0	0.089	G:A
673	rs6428388	195378717	0	0	1	100	30	0	0	T:T
674	rs12025066	195379068	0.017	0.017	1	97.8	28	0	0.009	T:C
675	rs6677082	195379156	0.161	0.148	1	90	22	0	0.08	A:G
676	rs1571964	195379696	0.161	0.148	1	90	22	0	0.08	A:G
677	rs6656858	195380196	0.161	0.148	1	90	22	0	0.08	T:C
678	rs7527211	195381081	0	0	1	93.3	24	0	0	A:A
679	rs4326657	195385567	0.033	0.033	1	100	30	0	0.017	A:G
680	rs4483441	195386412	0	0	1	100	30	0	0	C:C
681	rs10922171	195388926	0	0	1	100	30	0	0	G:G
682	rs1127661	195389666	0.446	0.481	0.7433	90	22	0	0.402	T:C
683	rs12031996	195391303	0.017	0.017	1	100	30	0	0.008	T:C
684	rs4639796	195393272	0.185	0.226	0.3944	87.8	21	0	0.13	G:A
685	rs12028827	195394114	0.179	0.219	0.3706	90	22	0	0.125	A:C
686	rs12724106	195394201	0.203	0.209	1	98.9	29	0	0.119	A:G
687	rs12128631	195395349	0.182	0.222	0.3822	88.9	21	0	0.127	G:A
688	rs12137359	195396604	0.179	0.219	0.3706	90	22	0	0.125	C:T
689	rs4244139	195397997	0.5	0.5	1	100	30	0	0.5	A:A
690	rs4342879	195398278	0.167	0.153	1	100	30	0	0.083	C:T
691	rs4350226	195399001	0.167	0.153	1	100	30	0	0.083	A:G
692	rs6677930	195402089	0.475	0.5	0.846	98.9	29	0	0.492	A:C
693	rs10732295	195403268	0.179	0.163	1	90	22	0	0.089	C:T
694	rs10732296	195403774	0.167	0.153	1	100	30	0	0.083	G:A
695	rs4520444	195408611	0.491	0.476	1	88.9	22	0	0.391	G:T
696	rs6656448	195409901	0.155	0.143	1	96.7	28	0	0.078	T:C
697	rs5029273	195412005	0.167	0.153	1	100	30	0	0.083	G:C
698	rs10429911	195412767	0.167	0.153	1	100	30	0	0.083	C:A
699	rs12037579	195415311	0	0	1	100	30	0	0	G:G
700	rs7513826	195417261	0.167	0.153	1	100	30	0	0.083	G:A
701	rs12046781	195417494	0	0	1	97.8	28	0	0	C:C
702	rs1415217	195417869	0.167	0.153	1	100	30	0	0.083	T:C

FIG. 14 CONT.

703	rs1556763	195418772	0.179	0.163	1	90	22	0	0.089	C:A
704	rs2039639	195419375	0	0	1	100	30	0	0	G:G
705	rs7520503	195419612	0.179	0.163	1	90	22	0	0.089	G:A
706	rs7523013	195419811	0.167	0.153	1	100	30	0	0.083	G:A
707	rs10922177	195420390	0.169	0.209	0.3387	98.9	29	0	0.119	T:C
708	rs7410943	195421644	0.483	0.5	0.9497	100	30	0	0.492	G:A
709	rs1415212	195424517	0.467	0.473	1	100	30	0	0.383	T:A
710	rs1415213	195424617	0	0	1	98.9	29	0	0	A:A
711	rs12746924	195426592	0	0	1	96.7	27	0	0	A:A
712	rs4915378	195427835	0.111	0.105	1	90	23	1	0.056	G:T
713	rs7531669	195427888	0	0	1	100	30	0	0	A:A
714	rs4915379	195428051	0.185	0.168	1	86.7	20	0	0.093	C:T
715	rs7542219	195428170	0	0	1	100	30	0	0	G:G
716	rs7534539	195428254	0	0	1	98.9	29	0	0	T:T
717	rs7534554	195428297	0	0	1	90	24	1	0	T:T
718	rs11801039	195429349	0	0	1	100	30	0	0	C:C
719	rs10494747	195430551	0.375	0.4	0.8173	90	22	0	0.277	T:A
720	rs10754218	195431637	0.375	0.4	0.8173	90	22	0	0.277	A:G
721	rs10754219	195431809	0.153	0.141	1	97.8	28	0	0.076	T:C
722	rs10801596	195431890	0.45	0.439	1	100	30	0	0.325	A:G
723	rs7516145	195432191	0	0	1	100	30	0	0	C:C
724	rs7516235	195432260	0	0	1	100	30	0	0	C:C
725	rs7516246	195432335	0.429	0.436	1	90	22	0	0.321	T:C
726	rs7530569	195432524	0	0	1	100	30	0	0	T:T
727	rs10922181	195432675	0.383	0.439	0.448	100	30	0	0.325	G:C
728	rs16841212	195433189	0	0	1	100	30	0	0	T:T
729	rs16841214	195433257	0	0	1	100	30	0	0	T:T
730	rs11808524	195433414	0	0	1	100	30	0	0	A:A
731	rs11808883	195433545	0	0	1	100	30	0	0	T:T
732	rs11800461	195433573	0	0	1	100	30	0	0	C:C
733	rs12402465	195434088	0.321	0.316	1	90	22	0	0.196	C:T
734	rs16841219	195434221	0	0	1	100	30	0	0	T:T
735	rs16841220	195434420	0	0	1	100	30	0	0	A:A
736	rs10494748	195434801	0	0	1	100	30	0	0	G:G
737	rs10494749	195434856	0	0	1	100	30	0	0	G:G
738	rs10494750	195434889	0	0	1	100	30	0	0	G:G
739	rs10494751	195435248	0.019	0.019	1	86.7	20	0	0.009	C:A
740	rs11811295	195436380	0	0	1	100	30	0	0	G:G
741	rs11801772	195436418	0	0	1	100	30	0	0	C:C

FIG. 14 CONT.

742	rs6690022	195436650	0.483	0.439	0.6753	100	30	0	0.325	G:A
743	rs12742517	195436921	0	0	1	100	30	0	0	T:T
744	rs12757081	195436922	0	0	1	98.9	29	0	0	C:C
745	rs7535048	195437805	0	0	1	98.9	29	0	0	C:C
746	rs16841237	195438689	0	0	1	100	30	0	0	C:C
747	rs12569003	195441706	0	0	1	100	30	0	0	G:G
748	rs6700604	195442216	0	0	1	86.7	19	0	0	G:G
749	rs6702739	195445655	0	0	1	100	30	0	0	A:A
750	rs7544067	195446333	0.483	0.439	0.6753	96.7	27	0	0.325	T:C
751	rs10922184	195446418	0.379	0.366	1	97.8	28	0	0.241	A:C
752	rs6428392	195446777	0	0	1	97.8	28	0	0	A:A
753	rs12026252	195447943	0.397	0.434	0.6588	96.7	27	0	0.319	G:T
754	rs12731997	195448561	0	0	1	100	30	0	0	A:A
755	rs12732003	195448569	0	0	1	95.6	26	0	0	A:A
756	rs12732005	195448572	0	0	1	88.9	21	0	0	A:A
757	rs10801598	195450279	0.404	0.438	0.7046	96.7	27	0	0.325	G:A
758	rs1415216	195452272	0.483	0.439	0.6753	100	30	0	0.325	T:C
759	rs1764627	195452824	0	0	1	100	30	0	0	A:A
760	rs12734982	195453985	0	0	1	100	30	0	0	A:A
761	rs10922187	195454228	0.464	0.436	0.9249	90	22	0	0.321	T:C
762	rs510135	195454402	0.167	0.153	1	100	30	0	0.083	T:C
763	rs613010	195454956	0	0	1	100	30	0	0	A:A
764	rs518149	195455287	0.169	0.155	1	98.9	29	0	0.085	G:A
765	rs615647	195455531	0.167	0.153	1	100	30	0	0.083	C:T
766	rs1764630	195458372	0	0	1	98.9	29	0	0	T:T
767	rs12125062	195459541	0.357	0.357	1	90	22	0	0.232	T:C
768	rs16841270	195459839	0	0	1	100	30	0	0	C:C
769	rs1890013	195461684	0.357	0.337	1	91.1	24	0	0.214	G:A
770	rs4915406	195462273	0.464	0.436	0.9249	90	22	0	0.321	T:C
771	rs11803081	195462283	0	0	1	100	30	0	0	G:G
772	rs524248	195465048	0.07	0.068	1	95.6	28	1	0.035	T:C
773	rs11586109	195465153	0	0	1	100	30	0	0	C:C
774	rs671076	195465559	0	0	1	100	30	0	0	G:G
775	rs590864	195469106	0	0	1	100	30	0	0	C:C
776	rs12730229	195469531	0	0	1	100	30	0	0	C:C
777	rs555406	195472803	0	0	1	100	30	0	0	A:A
778	rs553571	195473016	0	0	1	100	30	0	0	T:T
779	rs6428393	195473788	0	0	1	100	30	0	0	A:A
780	rs12128816	195474228	0.357	0.357	1	90	22	0	0.232	A:G

FIG. 14 CONT.

781	rs3861926	195474325	0.033	0.033	1	97.8	28	0	0.017	G:A
782	rs12757091	195474889	0.067	0.064	1	100	30	0	0.033	T:G
783	rs9427665	195476640	0.383	0.367	1	98.9	29	0	0.242	A:G
784	rs12082890	195479450	0	0	1	100	30	0	0	T:T
785	rs10922190	195480711	0.393	0.448	0.4811	90	22	0	0.339	A:G
786	rs12136929	195481400	0.373	0.362	1	98.9	29	0	0.237	T:G
787	rs1337165	195481465	0	0	1	100	30	0	0	A:A
788	rs1582118	195482296	0.375	0.366	1	95.6	26	0	0.241	T:C
789	rs10494752	195482842	0.393	0.448	0.4811	90	22	0	0.339	C:T
790	rs1764629	195483255	0.179	0.163	1	90	22	0	0.089	G:A
791	rs17616836	195483304	0.5	0.448	0.6238	90	22	0	0.339	G:A
792	rs10801600	195483589	0.418	0.458	0.6727	93.3	24	0	0.355	T:C
793	rs12119142	195487871	0.382	0.37	1	88.9	21	0	0.245	C:T
794	rs6678066	195488527	0.167	0.153	1	100	30	0	0.083	G:A
795	rs523427	195490070	0.033	0.033	1	100	30	0	0.017	G:A
796	rs4915202	195493617	0	0	1	91.1	23	0	0	G:G
797	rs517222	195493656	0.357	0.357	1	90	22	0	0.232	G:A
798	rs12402497	195494563	0.05	0.049	1	100	30	0	0.025	C:A
799	rs1764632	195495688	0	0	1	91.1	23	0	0	A:A
800	rs1613423	195496035	0	0	1	97.8	28	0	0	C:C
801	rs1613427	195496039	0	0	1	100	30	0	0	T:T
802	rs11810183	195496928	0	0	1	100	30	0	0	A:A
803	rs11810588	195497294	0	0	1	97.8	28	0	0	T:T
804	rs1764633	195498057	0	0	1	100	30	0	0	C:C
805	rs4915440	195498434	0.386	0.444	0.4406	96.7	27	0	0.333	C:G
806	rs6660018	195499478	0.383	0.367	1	98.9	29	0	0.242	G:A
807	rs476505	195499872	0	0	1	100	30	0	0	C:C

FIG. 14 CONT.

ID	Haplotype	Haplotype Sequence
NO.	H1_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTTGGCGGGCTTCGTCC
220	H2_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
230	H3_51_B	ACTAATCCTTTTCATAAGAGTGCGCGATACGGGGTGGCTGGTTTGGTCC
231	H4_51_B	ATCGATCCTTTACGAAGGGGCTTCGAATGCAAAGTAGATTATCTTCATCC
232	H5_51_B	ACCGGCTCTTTATAAAGATCGCGGATGCAAGTCTGATCATCTTACTT
233	H6_51_B	ACTAATCCTTTTCATAAGAGTGCGCGATGCAAGTAGATTATCTTCATCC
234	H7_51_B	GTCGATCCTTTATGAAAGGCCOCCGGACGGGGTGGGTATCTTCATCC
235	H8_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
236	H9_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTGCAGGGTTGGCGGGCTTCGTCC
237	H10_51_B	ATCGATCAACCATGACAAGGCOCGGACGGGGTGGCGGGCTTCGTCC
238	H11_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTGGGGGTTGGCGGGCTTCGTCC
239	H12_51_B	ATCGATCCTTTATAAAGATCGCGGAATGCAAGTAGATTATCTTCATCC
240	H13_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGCGGGCTTCGTCC
241	H14_51_B	ACCGGCTCTTTATAAAGATCGCGGATGCAAGTAGATTATCTTCATCC
242	H15_51_B	ACCGGCTCTTTATAAAGATCGCGGATGCAAGTAGATTATCTTCATCC
243	H16_51_B	ATCGGCTCTTTATAAAGATCGCGGATGCAAGTAGATTATCTTCATCC
244	H17_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
245	H18_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTACGGGGTGGGTATCTTCATCC
246	H19_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
247	H20_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
248	H21_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
249	H22_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
250	H23_51_B	ATCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
251	H24_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
252	H25_51_B	ATCGATCCTTTACGAAGGGGCTTCGAATGCAAGTAGATTATCTTCATCC
253	H26_51_B	ACTAATCCTTTACGAAGGGGCTTCGAATGCAAGTAGATTATCTTCATCC
254	H27_51_B	ACTAATCCTTTACGAAGGGGCTTCGAATGCAAGTAGATTATCTTCATCC
255	H28_51_B	ATCGATCCTTTACGAAGGGGCTTCGAAGGGGTTGGGTATCTTCATCC
256	H29_51_B	GTCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
257	H30_51_B	ATCGATCCTTTATAAAGATCGCGGATACGGGGTGGGTATCTTCATCC
258	H31_51_B	ACCGATCAACCATGACAAGGCOCGGACGGGGTGGGTATCTTCATCC
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H32_51_B	ATCGATCCTTTATATGAAGATCGCGGAATGCAAAAGTAGATATCTCATCC	A	T	C	G	A	T	C	C	T	T	A	T	A	A	G	A	T	C	C	G	A	T	G	C	A	A	A	G	T	A	T	C	T	C	C	A	T	C	C									
H33_51_B	ACCGATCCTTTACGAAGGGGCGCTTCGAATGCAAAAGTAGATATCTCATCC	A	C	C	G	A	T	C	C	T	T	A	C	G	A	G	G	G	C	C	T	T	C	G	A	T	G	C	A	A	G	T	A	T	C	T	T	C	A	T	C	C							
H34_51_B	ATCGATCCTTTATATGAAGATCGCGGAATGCAAAAGTTGCGGSGCTCGTCC	A	T	C	G	A	T	C	C	T	T	A	T	A	A	A	G	A	T	C	C	G	C	C	G	A	T	G	C	A	A	A	G	T	G	C	G	C	T	C	G	T	C	C					
H35_51_B	GTGATCAACCAATGACAAGGCGCGGGAAGCGGAGTTGGGTATCTCATCC	G	T	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	G	C	G	A	C	G	C	G	G	G	T	G	C	G	A	T	C	T	C	A	T	C	C						
H36_51_B	ACCGATCAACCAATGACAAGGCGCGGGAAGCGGAGTTGGGTATCTCATCC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	G	C	C	G	A	C	G	C	G	G	T	T	G	C	T	C	G	T	T	T	C	G	T	C	C				
H37_51_B	ACTAATCCTTTTATGAAGATCGCGGAATGCGGGGTTGGGTATCTCATCC	A	C	T	A	T	C	C	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G	A	T	G	C	G	G	G	T	G	C	G	T	A	T	C	T	C	A	T	C	C			
H38_51_B	ACTAATCCTTTACATAAGAGTGCGCGAATGACGGGGTTGCTCGTTTGGTCC	A	C	T	A	A	C	T	C	T	T	A	C	A	T	A	A	G	A	G	T	C	G	C	C	G	A	T	A	C	G	G	G	T	G	C	T	C	G	T	T	T	C	G	T	C	C		
H39_51_B	ACCGATCCTTTATGACAAGGCGCGGGAAGCGGAGTTGGGTATCTCATCC	A	C	C	G	A	T	C	C	T	T	A	T	G	A	C	A	A	G	C	C	G	C	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	C	T	C	A	T	C	C			
H40_51_B	ATCGGCTCTTTATATGAAGATCGCGGAGTGACGGGTTGCGGGCTCGTCC	A	T	C	G	C	T	C	T	T	A	T	A	A	A	G	A	T	C	C	G	C	C	G	A	T	G	C	A	G	G	T	T	G	C	G	C	G	T	T	C	G	T	C	C				
H41_51_B	ACTAATCCTTTTATGAAGATCGCGGAGTACGGGGTTGGGTATCTCATCC	A	C	T	A	T	C	C	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G	A	T	A	C	G	G	G	T	T	G	C	G	T	A	T	C	T	C	A	T	C	C		
H42_51_B	ACTAGCTCTTTATATGAAGATCGCGGAATGCAAAAGTAGATATCTCATCC	A	C	T	A	G	C	T	C	T	T	A	T	A	A	A	G	A	T	C	C	G	C	C	G	A	T	G	C	A	A	A	G	T	A	A	G	T	A	T	C	T	C	A	T	C	C		
H43_51_B	ACTAATCCTTTTATGAAGATCGCGGATACGGGGTTGCTCGGCTCGTCC	A	C	T	A	T	C	C	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G	A	T	A	C	G	G	G	T	T	G	C	T	C	G	C	T	T	C	G	T	C	C		
H44_51_B	GTGATCAACCAATGACAAGGCGCGGGAAGCGGAGTTGGGAGCTCGTCC	G	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	G	C	C	G	A	C	G	C	G	G	G	T	T	G	C	G	C	T	T	C	G	C	T	C	C				
H45_51_B	ACCGATCCTTTTATGAAGATCGCGGATACGGGGTTGCTCGTTTGGTCC	A	C	C	G	A	T	C	C	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G	A	T	A	C	G	G	G	T	T	G	C	T	C	G	T	T	T	C	G	T	C	C	
H46_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTGGGGTTGGGTATCTCATCC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	G	A	T	G	G	G	T	T	G	C	G	T	A	T	C	T	C	A	T	C	C			
H47_51_B	ATCGATCAACCAATGACAAGGCTTCGAATGCAAAAGTAGATATCTCATCC	A	T	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	T	T	C	G	A	T	G	C	A	A	A	G	T	A	A	G	T	A	T	C	T	C	A	T	C	C			
H48_51_B	ATCGATCCTTTATATGAAGATCCTTCGAGTGGGGTTGCGGSGCTCGTCC	A	T	C	G	A	T	C	C	T	T	A	T	A	A	A	G	A	T	C	C	T	T	C	G	A	T	G	C	G	G	G	T	T	G	C	G	C	T	T	C	G	T	C	C				
H49_51_B	ACTAATCAACCAATGACAAGGCGCGGGAAGCGGAGTTGGGAGCTCGTCC	A	C	T	A	T	C	A	C	A	C	A	T	G	A	C	A	A	G	C	C	G	C	C	G	A	C	G	C	G	G	T	T	G	C	G	C	T	T	C	G	T	C	C					
H50_51_B	ACCGATCAACCAATGACAAGGCGCGGGAAGCGGAGTTGGGTATCTTACTT	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	G	C	C	G	A	C	G	C	G	G	T	C	T	G	A	T	C	A	T	C	T	T	A	C	T	T			
H51_51_B	GTGATCCTTTTATGACAAGGCGCGCGGATACGGGGTTGCTCGTTTGGTCC	G	T	C	G	A	T	C	C	T	T	A	T	G	A	C	A	A	G	C	C	G	C	C	G	A	T	A	C	G	G	G	T	T	G	C	T	C	G	T	T	T	C	G	T	C	C		
H52_51_B	GTGATCAACCAATGACAAGGCGCGCGGAGCGGGGTTGGGTATCTCATCC	G	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	G	C	C	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	C	T	C	A	T	C	C			
H53_51_B	ACTAATCCTTTTATGAAGATGCGCGAGTGGGGGTTGGGAGCTGGTCC	A	C	T	A	T	C	C	T	T	T	C	A	T	A	A	A	G	T	C	G	C	C	G	A	C	G	C	G	G	G	T	T	G	C	G	C	G	T	T	C	G	T	C	C				
H54_51_B	ACCGATCCTTTACGAAGGGGCTTCGAGTGGGGTTGCGGGCTCGTCC	A	C	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	C	C	T	T	C	G	A	T	G	T	G	G	G	T	T	G	C	G	C	G	T	T	C	G	T	C	C				
H55_51_B	ATCGATCCTTTATATGAAGATCGCGGAATGCAAGGTTGGGTATCTCATCC	A	T	C	G	A	T	C	C	T	T	A	T	A	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	G	G	T	T	G	C	G	T	A	T	C	T	C	A	T	C	C			
H56_51_B	ATCGATCCTTTTATGAAGATGCGCGGATACGGGGTTGCTCGTTTGGTCC	A	T	C	G	A	T	C	C	T	T	T	C	A	T	A	A	A	G	A	G	T	C	G	C	C	G	A	T	A	C	G	G	G	T	T	G	C	T	C	G	T	T	T	C	G	T	C	C

FIG. 15 CONT.

Haplotype	Freq.	Case, Control Ratio Counts	Case, Control Freqs	Chi Square	P Value
H1_51_B	0.214	1240.3 : 3884.9, 313.5 : 1480.7	0.242, 0.175	34.545	4.17E-09
H2_51_B	0.15	901.0 : 4224.2, 189.5 : 1604.7	0.176, 0.106	49.31	2.19E-12
H3_51_B	0.084	360.8 : 4764.4, 246.5 : 1547.6	0.070, 0.137	74.515	6.02E-18
H4_51_B	0.069	364.5 : 4760.7, 136.9 : 1657.2	0.071, 0.076	0.536	0.4642
H5_51_B	0.051	228.6 : 4896.5, 140.0 : 1654.2	0.045, 0.078	29.424	5.81E-08
H6_51_B	0.038	192.6 : 4932.6, 85.0 : 1709.2	0.038, 0.047	3.306	0.069
H7_51_B	0.028	147.9 : 4977.3, 52.4 : 1741.7	0.029, 0.029	0.006	0.9372
H8_51_B	0.02	107.7 : 5017.4, 35.0 : 1759.2	0.021, 0.019	0.155	0.6937
H9_51_B	0.018	101.8 : 5023.3, 31.9 : 1762.3	0.020, 0.018	0.311	0.5768
H10_51_B	0.018	104.0 : 5021.2, 27.2 : 1767.0	0.020, 0.015	1.888	0.1694
H11_51_B	0.017	94.7 : 5030.5, 28.0 : 1766.1	0.018, 0.016	0.614	0.4333
H12_51_B	0.017	67.0 : 5058.2, 55.6 : 1738.6	0.013, 0.031	24.48	7.51E-07
H13_51_B	0.015	91.4 : 5033.8, 16.0 : 1778.2	0.018, 0.009	6.911	0.0086
H14_51_B	0.013	48.6 : 5076.6, 46.9 : 1747.3	0.009, 0.026	27.149	1.88E-07
H15_51_B	0.012	53.4 : 5071.7, 32.7 : 1761.5	0.010, 0.018	6.563	0.0104
H16_51_B	0.011	54.2 : 5071.0, 28.8 : 1765.4	0.011, 0.016	3.362	0.0667
H17_51_B	0.011	70.5 : 5054.7, 11.3 : 1782.9	0.014, 0.006	6.348	0.0118
H18_51_B	0.011	59.8 : 5065.3, 17.9 : 1776.3	0.012, 0.010	0.355	0.5514
H19_51_B	0.01	54.3 : 5070.9, 17.2 : 1776.9	0.011, 0.010	0.126	0.7222
H20_51_B	0.01	47.9 : 5077.3, 23.4 : 1770.8	0.009, 0.013	1.78	0.1822
H21_51_B	0.01	49.6 : 5075.6, 19.8 : 1774.4	0.010, 0.011	0.236	0.6269
H22_51_B	0.01	55.9 : 5069.3, 13.0 : 1781.1	0.011, 0.007	1.782	0.1819
H23_51_B	0.009	54.0 : 5071.2, 10.4 : 1783.8	0.011, 0.006	3.264	0.0708
H24_51_B	0.008	50.0 : 5075.2, 8.0 : 1786.1	0.010, 0.004	4.426	0.0354
H25_51_B	0.006	31.7 : 5093.5, 14.4 : 1779.7	0.006, 0.008	0.696	0.4042
H26_51_B	0.006	28.9 : 5096.3, 16.0 : 1778.1	0.006, 0.009	2.246	0.134
H27_51_B	0.006	32.2 : 5092.9, 10.8 : 1783.3	0.006, 0.006	0.013	0.9099
H28_51_B	0.006	37.6 : 5087.6, 3.4 : 1790.7	0.007, 0.002	6.612	0.0101
H29_51_B	0.006	29.5 : 5095.7, 11.2 : 1783.0	0.006, 0.006	0.054	0.8164
H30_51_B	0.005	22.0 : 5103.2, 17.7 : 1776.5	0.004, 0.010	7.22	0.0072
H31_51_B	0.005	30.6 : 5094.6, 8.4 : 1785.8	0.006, 0.005	0.394	0.5301

FIG. 15 CONT.

H32_51_B	0.005	24.8 : 5100.4, 12.2 : 1782.0	0.005, 0.007	0.963	0.3264
H33_51_B	0.004	20.3 : 5104.8, 11.0 : 1783.2	0.004, 0.006	1.37	0.2418
H34_51_B	0.004	21.9 : 5103.3, 8.0 : 1786.2	0.004, 0.004	0.012	0.9112
H35_51_B	0.004	23.7 : 5101.4, 5.0 : 1789.2	0.005, 0.003	1.095	0.2953
H36_51_B	0.004	19.5 : 5105.6, 9.2 : 1785.0	0.004, 0.005	0.544	0.4607
H37_51_B	0.004	19.5 : 5105.6, 7.7 : 1786.5	0.004, 0.004	0.074	0.7857
H38_51_B	0.003	19.0 : 5106.2, 6.0 : 1788.2	0.004, 0.003	0.049	0.8254
H39_51_B	0.003	18.8 : 5106.4, 6.0 : 1788.2	0.004, 0.003	0.045	0.8314
H40_51_B	0.003	12.0 : 5113.2, 9.1 : 1785.1	0.002, 0.005	3.216	0.0729
H41_51_B	0.003	16.0 : 5109.2, 4.8 : 1789.4	0.003, 0.003	0.091	0.7635
H42_51_B	0.003	13.0 : 5112.2, 6.0 : 1788.2	0.003, 0.003	0.316	0.5743
H43_51_B	0.002	8.1 : 5117.1, 8.9 : 1785.3	0.002, 0.005	6.221	0.0126
H44_51_B	0.002	13.3 : 5111.9, 2.0 : 1792.1	0.003, 0.001	1.287	0.2566
H45_51_B	0.002	9.4 : 5115.8, 4.0 : 1790.1	0.002, 0.002	0.118	0.7312
H46_51_B	0.002	7.2 : 5118.0, 4.2 : 1790.0	0.001, 0.002	0.72	0.3961
H47_51_B	0.001	7.2 : 5118.0, 3.0 : 1791.2	0.001, 0.002	0.059	0.8074
H48_51_B	0.001	10.0 : 5115.2, 0.0 : 1794.2	0.002, 0.000	3.506	0.0612
H49_51_B	0.001	6.7 : 5118.4, 3.2 : 1791.0	0.001, 0.002	0.194	0.6595
H50_51_B	0.001	7.1 : 5118.1, 2.2 : 1792.0	0.001, 0.001	0.024	0.8772
H51_51_B	0.001	6.2 : 5119.0, 2.7 : 1791.4	0.001, 0.002	0.101	0.7501
H52_51_B	0.001	7.8 : 5117.3, 1.0 : 1793.1	0.002, 0.001	0.938	0.3327
H53_51_B	0.001	5.9 : 5119.3, 1.5 : 1792.6	0.001, 0.001	0.105	0.7465
H54_51_B	0.001	5.4 : 5119.8, 2.0 : 1792.2	0.001, 0.001	0.003	0.9531
H55_51_B	0.001	4.3 : 5120.9, 3.1 : 1791.1	0.001, 0.002	0.946	0.3308
H56_51_B	0.001	4.8 : 5120.4, 2.5 : 1791.7	0.001, 0.001	0.247	0.6193

FIG. 15 CONT.

[illegible]

FIG. 16

[illegible]

FIG. 17

31	H31_62_A	ATCSAATC0ATATN4ATAGTGA4GCG0000CCGAAAGTGGGCTAGCTGGTAAAGCAAG	A	T	C	G	S	A	T	C	C	A	T	A	T	A	T	M	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G				
32	H32_62_A	ATCSAATC0ATATN4AGATGCGGGAATGATT03A4G6G6GCGTACCGAATCAAGTAAGG	A	T	C	G	S	A	T	C	C	A	T	A	T	A	T	N	G	A	T	G	G	A	T	G	G	A	C	T	T	G	A	T	T	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G			
33	H33_62_A	ACTAJATC0ATATN4TGATAGTGA4GCG04GCGAAGATGGGCTAGCGGGGCAAGCAAGG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	G	C	A	T	G	A	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	G	G	G	C	A	G	C	A	C	C	G				
34	H34_62_A	ACTAJATC0ATATN4TGATAGTGA4GCG04GCGAAGATGGGCTAGCGGGGCAAGCAAGG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	G	C	A	T	G	A	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	C	A	G	C	A	C	C	G				
35	H35_62_A	AC0GSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	C	G	T	A	T	C	G	G	G	C	A	G	C	A	C	C	G			
36	H36_62_A	AC0GSAATC0CA4TNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	S	A	T	C	C	A	T	A	T	N	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	A	T	C	A	G	A	T	C	A	C	C	G			
37	H37_62_A	ACTAJATC0ATATN4TGATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	G	G	C	A	G	C	A	C	C	G				
38	H38_62_A	ATCSAATC0ATATN4ATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	T	C	G	S	A	T	C	C	A	T	A	T	A	T	M	A	A	T	A	G	T	G	A	C	T	T	G	A	T	T	C	C	A	G	G	C	G	G	C	G	T	A	C	C	G	G	G	C	A	G	C	A	C	C	G					
39	H39_62_A	ATCSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	T	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	A	T	C	A	G	A	T	C	A	C	C	G			
40	H40_62_A	ACTAJATC0ATATN4TGATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	C	A	T	G	A	C	C	G	A	A	G	C	G	C	G	T	A	C	C	G	A	T	C	A	G	A	T	C	A	C	C	G				
41	H41_62_A	AC0SAATC0ATATN4TGATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	C	A	T	G	A	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G				
42	H42_62_A	ATCSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	T	C	G	S	A	T	C	A	T	C	G	C	A	G	D	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	A	T	C	A	G	A	T	C	A	C	C	G	
43	H43_62_A	AC0GSAATC0CA4TNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	S	A	T	C	A	T	C	G	C	A	T	A	G	N	A	A	T	A	G	T	G	A	C	C	A	T	G	A	C	C	G	A	G	G	C	A	G	C	T	C	A	T	G	A	T	C	A	T	G	A	T	C	A	T	G	T	A
44	H44_62_A	AC0GSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	C	T	C	A	T	G	A	T	C	A	A	T	G	T	A					
45	H45_62_A	AC0GSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G			
46	H46_62_A	AC0GSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	A	C	G	C	C	G	C	D	D	D	C	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	A	T	C	A	G	A	T	C	A	C	C	G		
47	H47_62_A	GTCSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	G	T	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	A	T	C	A	G	A	T	C	A	C	C	G	
48	H48_62_A	ATCSAATC0ATATN4ATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	T	C	G	S	A	T	C	C	A	T	A	T	N	G	A	T	A	G	T	G	G	A	C	T	T	G	A	T	T	C	C	A	A	G	C	A	G	C	G	T	A	C	C	G	G	G	C	A	G	C	A	C	C	G						
49	H49_62_A	ATCSAATC0ATATN4ATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	T	C	G	S	A	T	C	C	A	T	A	T	N	A	A	T	A	G	T	T	G	A	C	G	C	D	D	D	C	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	G	G	C	A	G	C	A	C	C	G						
50	H50_62_A	ATCSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	T	C	G	S	A	T	C	A	T	C	G	C	A	G	D	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	G	G	C	A	G	C	A	C	C	G			
51	H51_62_A	ACTAJATC0ATATN4TGATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	G	C	A	T	T	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G				
52	H52_62_A	ACTAJATC0ATATN4TGATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	G	C	A	T	T	C	C	G	A	A	G	T	G	A	T	G	T	C	A	T	A	A	T	C	A	G	A	T	C	A	C	C	G			
53	H53_62_A	GTCSAATC0ATATN4AG4AG4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	G	T	C	G	S	A	T	C	C	A	T	A	T	N	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G				
54	H54_62_A	ATCSAATC0ATATN4ATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	T	C	G	S	A	T	C	C	A	T	A	T	A	T	M	A	A	T	A	G	T	G	A	C	C	A	T	G	A	C	C	A	A	G	C	A	A	G	C	A	T	G	T	C	A	T	C	A	G	A	T	C	A	C	C	G				
55	H55_62_A	AC0GSAATC0ATATN4TGATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	S	A	T	C	C	A	T	A	T	N	G	A	T	A	T	A	G	T	G	G	A	C	T	T	G	A	T	T	C	C	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G				
56	H56_62_A	AC0GSAATC0CA4TNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	C	G	S	A	T	C	A	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	C	D	D	D	C	C	G	A	A	G	C	A	G	C	G	T	A	C	C	G	G	G	C	A	G	C	A	C	C	G				
57	H57_62_A	AC0GSAATC0ATATN4ATAGTGA4GCG0000CCGAAAGTGGGCTAGCTGGTAAAGCAAGG	A	C	C	G	S	A	T	C	C	A	T	A	T	N	A	A	T	A	T	A	G	T	G	A	C	C	D	D	D	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G					
58	H58_62_A	ACTAJATC0ATATN4TGATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	G	T	A	G	T	G	A	C	G	C	A	T	G	A	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G			
59	H59_62_A	ATCSAATC0CA4GNA4G4AGCGGCGATGA4GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	T	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	A	C	G	C	C	G	C	A	T	G	A	G	C	G	A	G	G	C	G	G	C	G	T	A	C	C	G	A	T	C	A	G	A	T	C	A	C	C	G	
60	H60_62_A	AC0GSAATC0ATATN4ATAGTGA4GCG0000CCGAAAGTGGGCTAGCTGGTAAAGCAAGG	A	C	C	G	S	A	T	C	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	C	D	D	D	C	C	G	A	G	G	C	G	C	G	T	A	C	C	G	A	T	C	A	G	A	T	C	A	C	C	G					
61	H61_62_A	ACTAJATC0ATATN4TGATAGTGA4GCG04GCGAAGGCGGGGATATCGGGGCAAGCAAGG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	C	A	T	G	A	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G				

FIG. 17 CONT.

62	H52_62_A	ACOSATCCATCCGACNAGAGAGACGGCCGATGACCGAGGGCCGCGCTACCGGGGCAAGTAACCG	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	A	G	T	A	C	C	G														
63	H53_62_A	ACOSATCCATCCGACNAGAGAGACGGCCGATGACCGAGGGCCGCGCTACCGGGGCAAGTAACCG	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	G	G	C	A	G	T	A	C	C	G														
64	H54_62_A	ACTAATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	N	G	G	A	T	T	A	G	G	C	G	G	A	C	T	T	G	A	T	T	C	C	G	A	G	G	C	A	G	T	A	C	C	G												
65	H55_62_A	ACTAATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	N	G	G	A	T	T	A	G	G	C	G	G	A	C	T	T	G	A	T	T	C	C	G	A	G	G	C	A	G	T	A	C	C	G												
66	H56_62_A	ACTAATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	N	G	G	A	T	T	A	G	G	C	G	G	A	C	T	T	G	A	T	T	C	C	G	A	G	G	C	A	G	T	A	C	C	G												
67	H57_62_A	ATTAAATCCATATCCTATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	T	A	L	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	C	C	G	A	A	G	T	G	G	C	T	A	C	T	G	T	A	A	G	C	A	C	C	G					
68	H58_62_A	ACOSATCCATCCGACNAGAGAGACGGCCGATGACCGAGGGCCGCGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	C	G	A	C	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	G	C	A	G	T	A	C	C	G										
69	H59_62_A	ATCSATCCATATCCTATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	T	C	G	S	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	G	C	T	A	C	T	G	T	A	A	G	C	A	C	C	G		
70	H70_62_A	ACTAATCCATATCCTATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	C	C	G	A	A	G	G	C	G	C	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G			
71	H71_62_A	GTCSATCCATATCCTATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	G	T	C	G	S	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	A	C	G	C	G	C	A	T	T	G	A	C	C	G	A	A	G	G	C	G	C	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G	
72	H72_62_A	GTCSATCCATATCCTATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	G	T	C	G	S	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	A	C	G	C	G	C	A	T	T	G	A	C	C	G	A	A	G	G	C	G	C	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G	
73	H73_62_A	ACOSATCCATCCGACNAGAGAGACGGCCGATGACCGAGGGCCGCGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	C	G	C	G	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	G	C	T	A	C	T	G	T	A	A	G	C	A	C	C	G			
74	H74_62_A	ATCSATCCATATCCTATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	T	C	G	S	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	C	A	T	G	T	A	A	G	T	A	C	C	G					
75	H75_62_A	ACOSATCCATCCGACNAGAGAGACGGCCGATGACCGAGGGCCGCGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	C	G	C	G	C	G	C	A	T	T	G	A	C	C	G	A	A	G	G	C	G	C	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G		
76	H76_62_A	ACOSATCCATCCGACNAGAGAGACGGCCGATGACCGAGGGCCGCGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	C	G	C	G	C	G	C	A	T	T	G	A	C	C	G	A	A	G	G	C	G	C	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G		
77	H77_62_A	ACTAATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	G	C	A	T	G	T	A	A	G	T	A	C	C	G						
78	H78_62_A	GTCSATCCATATCCTATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	G	T	C	G	S	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	C	A	T	G	T	A	A	G	T	A	C	C	G					
79	H79_62_A	ACTAATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	T	A	S	A	T	C	A	T	A	T	A	T	A	G	N	A	A	T	T	A	G	T	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	G	C	A	G	C	T	C	A	T	G	A	T	G	T	A					
80	H80_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	C	A	T	A	T	A	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	G	C	A	T	T	G	A	C	C	G	A	A	G	G	C	G	C	T	A	C	C	G	A	G	C	A	C	C	G		
81	H81_62_A	ACTAATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	G	C	T	A	C	C	G	A	G	C	A	C	C	G		
82	H82_62_A	ACTAATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	T	A	L	G	C	T	C	A	T	A	T	A	T	A	G	N	A	A	T	T	A	G	T	T	G	A	C	G	C	D	D	D	C	C	A	A	G	G	C	A	A	T	G	T	C	A	T	A	A	G	T	A	C	C	G		
83	H83_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	G	C	T	C	A	T	A	T	A	T	A	T	A	G	N	A	A	T	T	A	G	T	T	G	A	C	G	C	D	D	D	C	C	A	A	A	G	G	C	A	A	T	G	T	C	A	T	A	A	G	T	A	C	C	G
84	H84_62_A	ACOSATCCATCCGACNAGAGAGACGGCCGATGACCGAGGGCCGCGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	G	A	C	G	C	G	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	G	C	T	A	C	C	G	A	G	C	A	C	C	G					
85	H85_62_A	ATCSATCCATATCCTATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	T	C	G	S	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	C	T	T	G	A	T	T	C	C	A	A	G	G	C	A	A	T	G	T	C	A	A	G	T	A	C	C	G			
86	H86_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	C	A	T	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	G	C	T	A	C	C	G	A	G	C	A	C	C	G			
87	H87_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	C	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	A	T	G	T	C	A	T	A	A	G	T	A	C	C	G				
88	H88_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	C	A	T	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	G	C	T	A	C	C	G	A	G	C	A	C	C	G			
89	H89_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	C	A	T	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	A	T	T	G	A	C	C	G	A	A	G	T	G	A	T	G	T	C	A	T	A	A	G	T	A	C	C	G		
90	H90_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	C	A	T	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	T	G	A	T	G	T	C	A	T	A	A	G	T	A	C	C	G				
91	H91_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	A	T	C	A	T	G	C	A	T	T	N	G	G	A	T	T	A	G	T	T	G	A	C	G	C	T	T	G	A	T	T	C	C	G	A	A	G	C	A	A	T	G	T	C	A	A	G	T	A	C	C	G			
92	H92_62_A	ACOSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	C	G	S	G	C	T	C	A	T	A	T	A	T	A	T	A	G	N	A	A	T	T	A	G	T	T	G	C	G	C	D	D	D	C	C	G	A	A	G	T	G	A	T	C	A	T	G	A	T	C	A	A	G	T	A	C	C	G
93	H93_62_A	ATCSATCCATATATNGGATAGGCGGACCTTGATTCGAGGGCGGGCTACCGGGGCAAGTAACCG	A	T	C	G	S	A	T	C	C	A	T	A	T	A	T	T	N	G	G	A	T	T	A	G	T	G	A	C	G	C	C	T	T	G	A	T	T	C	C	A	G	A	G	G	C	A	G	C	T	A	C	C	G						

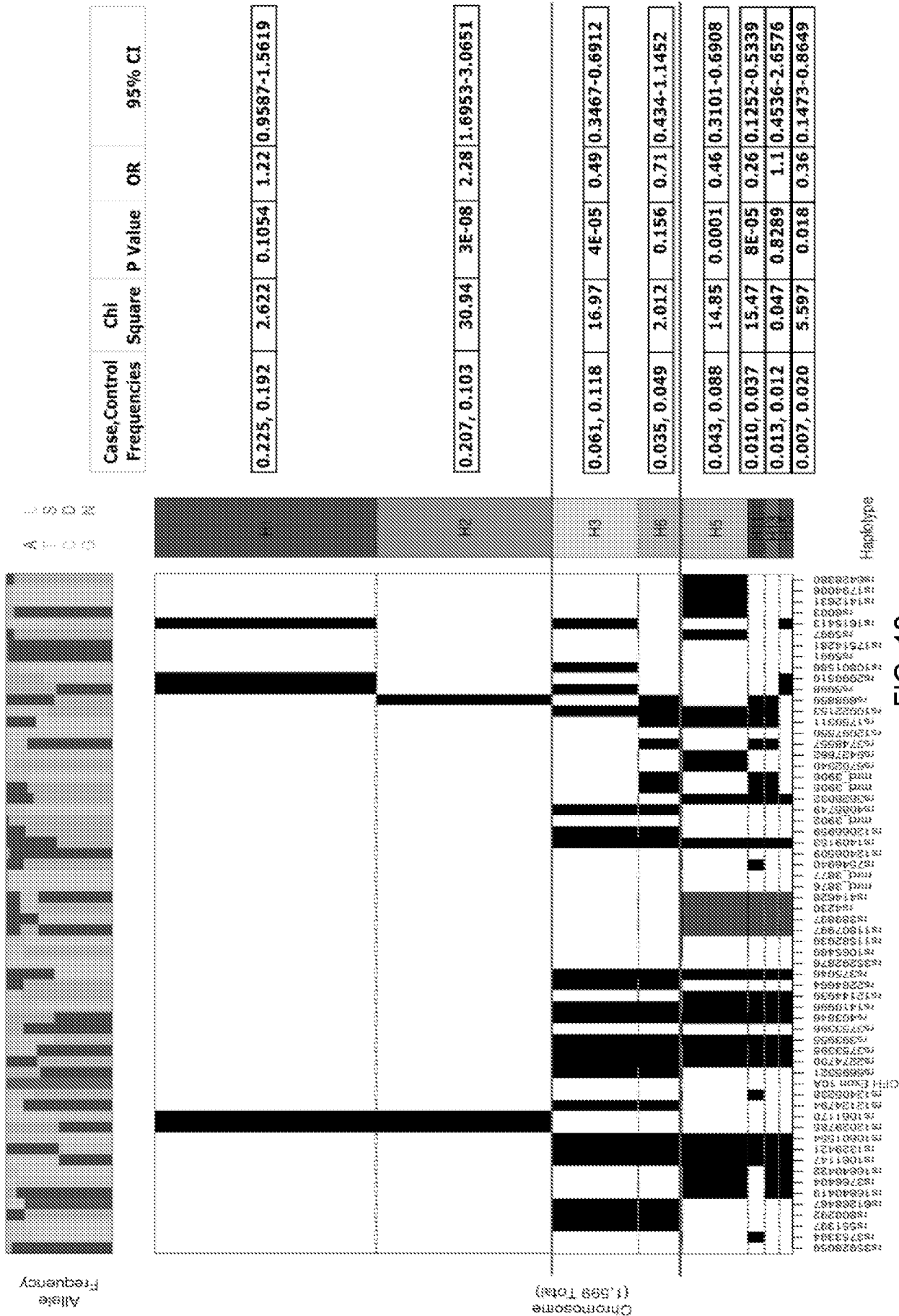
FIG. 17 CONT.

94	H94_62_A	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	A	A	C	G	C	C	A	T	G	A	C	C	G	A	G	G	C	G	T	A	C	C	G	G	G	C	A	A	G	A	C	C	G											
95	H95_62_A	A	C	C	G	S	A	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G				
96	H96_62_A	A	T	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	A	G	T	A	C	C	G		
97	H97_62_A	A	C	C	G	S	A	T	C	C	A	T	A	T	A	M	G	G	A	T	G	G	C	G	A	C	T	T	G	A	T	T	C	C	G	A	A	G	A	C	G	C	G	T	A	C	C	G	G	G	C	A	A	G	C	C	G			
98	H98_62_A	A	C	C	G	S	A	T	C	A	T	A	T	A	G	N	A	G	T	G	T	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G			
99	H99_62_A	A	T	C	G	S	A	T	C	C	A	T	A	T	A	M	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	G	C	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G			
100	H100_62_A	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	G	T	A	G	T	G	A	A	C	G	C	A	T	G	A	C	C	G	A	A	G	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G				
101	H101_62_A	A	T	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	G	C	G	T	A	C	C	G	G	G	C	A	A	G	A	C	C	G					
102	H102_62_A	A	C	C	G	L	G	C	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G		
103	H103_62_A	A	C	T	A	S	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	A	C	G	C	A	T	G	A	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G
104	H104_62_A	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	A	C	G	G	A	C	G	C	A	T	G	A	C	C	G	A	A	G	G	C	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G	
105	H105_62_A	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	M	G	A	T	A	G	T	G	A	A	C	G	C	A	T	G	A	C	C	G	A	A	G	G	C	A	T	C	A	T	G	A	T	G	A	T	G	T	A					
106	H106_62_A	A	C	T	A	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	G	C	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G		
107	H107_62_A	A	C	C	G	S	G	T	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	G	C	A	A	T	G	A	T	C	A	A	T	G	T	A	C	C	G					
108	H108_62_A	G	C	C	G	S	A	T	C	C	A	T	A	T	A	M	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	G	C	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G			
109	H109_62_A	A	T	C	G	S	A	T	C	C	A	T	A	T	A	M	G	A	T	G	G	C	G	A	C	T	T	G	A	T	T	C	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G					
110	H110_62_A	A	T	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	G	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	G	C	A	T	C	A	T	G	A	T	G	A	T	G	T	A	C	C	G				
111	H111_62_A	A	T	C	G	S	A	T	C	C	A	T	A	T	A	M	A	G	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G			
112	H112_62_A	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	A	C	G	C	T	G	C	A	T	G	A	C	A	A	G	A	G	G	C	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G		
113	H113_62_A	A	C	C	G	S	A	T	C	A	T	G	C	A	G	N	A	G	T	A	G	T	G	C	C	G	A	T	T	C	C	A	A	G	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G					
114	H114_62_A	A	C	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	G	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	G	C	T	C	A	T	G	A	T	G	A	T	G	T	A	C	C	G			
115	H115_62_A	A	T	C	G	S	A	T	C	C	A	T	A	T	A	M	A	A	T	A	G	T	G	A	C	G	C	A	T	G	A	C	C	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	G	T	A	C	C	G			
116	H116_62_A	A	T	C	G	S	A	T	C	C	A	T	A	T	A	M	G	G	A	T	G	G	C	G	A	C	T	T	G	A	T	T	C	C	G	A	G	G	C	G	C	G	T	A	C	C	G	G	G	C	A	A	G	C	A	C	C	G		
117	H117_62_A	A	T	C	G	S	A	T	C	C	A	T	A	T	A	M	G	G	A	T	G	G	C	G	A	C	T	T	G	A	G	A	C	C	G	A	A	G	G	C	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G	
118	H118_62_A	A	T	C	G	S	A	T	C	C	A	T	A	T	A	M	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	A	A	G	C	A	A	G	C	A	G	C	G	T	A	C	C	G	G	G	C	A	A	G	C	A	C	C	G
119	H119_62_A	A	C	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	G	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G			
120	H120_62_A	A	C	C	G	S	A	T	C	A	T	C	G	C	A	T	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	G	C	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G			
121	H121_62_A	G	T	C	G	S	A	T	C	C	A	T	A	T	A	M	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	A	G	T	G	G	C	G	T	A	C	C	T	G	G	T	A	A	G	C	A	C	C	G	
122	H122_62_A	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	T	G	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G		
123	H123_62_A	A	C	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	A	T	G	C	C	G	C	A	T	G	A	C	C	A	A	G	C	A	A	G	C	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G			
124	H124_62_A	A	C	C	G	S	A	T	C	C	A	T	A	T	A	G	N	G	A	T	A	G	T	G	A	C	G	C	A	T	G	A	C	C	G	A	A	G	G	C	G	T	A	C	C	G	A	A	T	C	A	G	T	A	C	C	G			
125	H125_62_A	A	C	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	G	C	T	C	A	T	A	A	T	C	A	A	G	T	A	C	C	G			

FIG. 17 CONT.

126	H126_62_A	ATCGSATCCHATTGNAGTAGTGAATGATTCGAAGGCAATGTTCAATCAAGTACCG	A	T	C	G	S	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	A	C	T	T	G	A	A	C	T	T	C	C	A	A	A	G	G	C	A	A	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G	
127	H127_62_A	ATCGSATCATCGCAGNAGATAGGAGCAAGCAATTCGAAGCGCGCTACTATGATCAAGTACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	T	A	G	C	G	G	A	C	G	C	A	A	T	T	C	C	A	A	A	G	G	C	G	C	T	T	C	A	T	G	A	T	C	A	G	T	A	C	C	G		
128	H128_62_A	ATCGSATCATCGCAGNAGAGAGCCGATGACGAAGAGCGCAATGTTCAATCAAGTACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	T	A	C	G	C	C	G	C	A	T	G	A	C	C	A	A	A	G	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	G	T	A	C	C	G		
129	H129_62_A	ATCGSATCATCGCAGNAGAGAGCCGATGACGAAGAGCGCGTACCGAATCAAGTACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	T	A	C	G	C	C	G	C	A	T	G	A	C	C	A	A	A	G	G	C	G	C	G	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G				
130	H130_62_A	ATCGS6CTCATAGNAATGATGAGTGGTACCGTCCDCCGAGAGCGCTTTCATCAAGTACCG	A	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	G	C	A	G	C	T	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G		
131	H131_62_A	ATCTAJATCCATATGNAATGATGAATGATTCGAGAGCAGCGCTTCATCAAGTACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	A	C	T	T	G	A	T	T	C	C	A	A	G	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G		
132	H132_62_A	ATCTALGCTCATATAGNAAGATGATGAGCCDCCGAAGCGCGCTCAAGTCAAGTACCG	A	C	T	A	L	G	C	T	C	A	T	A	T	A	G	N	A	G	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	G	C	G	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G				
133	H133_62_A	ATCTASATCATATGNGTGTAGTGAAGCATGAAGAGAGCTCACTGATCAAGTGTTA	A	C	T	A	S	A	T	C	C	A	T	A	T	T	G	N	G	G	T	A	G	T	G	A	A	C	G	C	A	T	G	A	C	C	G	A	A	G	C	A	G	C	T	C	A	C	A	T	G	A	T	C	A	A	T	G	T	A		
134	H134_62_A	ATCTALATCCATATTGAGGAAGAGCGCGATGAAGAGAGCGCGCTACCGAATCAAGTACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	G	C	G	C	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G			
135	H135_62_A	ATCGSATCCHATTATNAATATGATGAGCGAATTCGAAGGCAATGTTCAATCAAGTACCG	A	T	C	G	S	A	T	C	C	A	T	A	T	A	T	N	A	A	T	A	G	T	G	A	C	G	C	G	A	T	T	C	C	A	A	G	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G			
136	H136_62_A	ATCGSATCCHATTATNAATATGATGAGCCDCCGAAGCGCGCTACCGAATCAAGTACCG	A	T	C	G	S	A	T	C	C	A	T	A	T	A	T	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	A	A	G	C	G	C	G	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G						
137	H137_62_A	ATCGS6CCATCCAGNAGAGAGCGCATGAAGAGAGCGCGCTACCGG6GCAAGCAGCG	A	C	G	S	G	C	A	T	C	G	C	A	G	N	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	C	G	A	G	G	C	G	T	A	C	C	G	G	C	A	A	G	C	A	G	C	C	G			
138	H138_62_A	ATCGSATCCHATTATNGATGCGG6ACTTGAGCAAGAGAGCGG08TACCG66GCAAGCAGCG	A	T	C	G	S	A	T	C	C	A	T	A	T	T	N	G	G	A	T	G	C	G	G	A	C	T	T	G	A	C	C	G	A	A	C	C	G	A	A	G	A	C	G	G	C	T	A	C	C	G	G	C	A	A	G	C	C	G		
139	H139_62_A	ATCGS6CTCATAGNAATGATGAGCCDCCGAAGCGCGCTCACTGATCAAGTACCG	A	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	G	C	A	G	C	T	C	A	T	G	A	T	C	A	G	T	A	C	C	G			
140	H140_62_A	ATCGS6ATCATCGCAGNAGAGAGCCGATGACGAAGAGCGCGCTACCGAATCAAGTACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	C	G	A	G	G	C	G	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G			
141	H141_62_A	ATCGSATCCHATTAGNAATGATGAGCCDCCGAGAGCGACTCACTGATCAAGTGTTA	A	T	C	G	S	A	T	C	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	G	C	A	G	C	T	C	A	T	G	A	T	C	A	A	T	G	T	A			
142	H142_62_A	ATCGSATCATCGCAGNAGAGAGCCGATGACGAAGAGCGCGCTACCGAATCAAGTACCG	G	T	C	G	S	A	T	C	A	T	C	G	C	A	G	N	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	A	A	A	G	C	A	A	A	G	C	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G
143	H143_62_A	ATCGSATCCHATTATNAATATGATGAGCCGAAGAGCAATGTCACCG66GCAAGCAGCG	A	T	C	G	S	A	T	C	C	A	T	A	T	A	T	N	A	A	T	A	G	T	G	A	C	G	C	A	T	G	A	C	C	A	A	A	G	C	A	A	A	G	C	A	T	G	T	C	A	T	A	A	T	C	A	G	C	C	G	
144	H144_62_A	ATCGS6CTCATAGNAATGAGCG6ACTTGATTCGAAGGCAATGTTCAATCAAGTACCG	A	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	A	T	T	G	C	G	A	C	T	T	G	A	T	T	C	C	A	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G				
145	H145_62_A	ATCTALATCATATATAGAGAGAGCGCGCATGACCGAGAGCGG08TACCGAATCAAGTACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	N	A	G	A	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	C	G	C	G	C	G	T	A	C	G	A	T	C	A	G	T	A	C	C	G					
146	H146_62_A	ATCGSATCCHATTATNGATGCGG6ACTTGAGCAAGAGAGCGCTCACTGATCAAGTACCG	A	T	C	G	S	A	T	C	C	A	T	A	T	N	G	A	T	G	C	G	A	C	G	A	C	G	C	A	T	G	A	C	C	A	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G			
147	H147_62_A	ATCGSATCATGATNGAGTGG08GACTTGATTCGAAGGCAATGTTCAATCAAGTACCG	A	C	G	S	A	T	C	A	T	G	C	A	T	T	G	C	A	T	T	G	C	G	A	C	T	T	G	A	T	T	C	C	A	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G				
148	H148_62_A	ATCGS6CTCATAGNAATGATGAGCCDCCGAAGGCAAGTCACTGATCAAGTGTTA	A	C	G	S	G	C	T	C	A	T	A	T	A	G	N	A	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	G	C	A	G	C	T	C	A	T	G	A	T	C	A	A	T	G	T	A				
149	H149_62_A	ATCGSATCATCGCAGNAGAGAGCCGATGACGAAGAGCGG08TACCGG6GCAAGCAGCG	A	C	G	S	A	T	C	A	T	C	G	C	A	N	A	A	A	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	C	G	C	G	C	G	T	A	C	C	G	G	C	T	A	C	G	A	C	C	G				
150	H150_62_A	ATCTALATCATCGCAGNAGAGAGCCGATGACGAAGAGCGCGCTACCGAATCAAGTACCG	A	C	T	A	L	A	T	C	C	A	T	C	G	C	A	N	A	G	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	A	G	C	G	A	G	C	G	C	G	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G
151	H151_62_A	ATCTAJATCATATGNGTGTAGTGAAGCCDCCGAGAGAGCGACTCACTGATCAAGTGTTA	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	C	G	C	D	D	D	C	C	G	A	A	G	C	A	A	T	G	T	C	A	T	G	A	T	C	A	A	T	G	T	A					
152	H152_62_A	ATCTASATCATATAGNAAGATGATTCGAGAGAGCGCTCACTGATCAAGTGTTA	A	C	T	A	S	A	T	C	C	A	T	A	T	N	G	G	A	T	A	G	C	G	A	C	T	A	T	T	C	C	G	A	A	G	C	A	A	T	G	T	C	A	T	G	A	T	C	A	A	T	G	T	A							
153	H153_62_A	ATCTALATCATATATAGGATGCGG6ACTTGATTCGAAGGCAATGTTCAATCAAGTACCG	A	C	T	A	L	A	T	C	C	A	T	A	T	N	G	G	A	T	G	C	G	A	C	T	T	G	A	T	T	C	C	A	A	A	G	C	G	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G					
154	H154_62_A	ATCGSATCCHATTATNGATGCGG6ACTTGAGCAAGAGAGCGTACCG66GCAAGCAGCG	A	T	C	G	S	A	T	C	C	A	T	A	T	N	G	G	A	T	G	C	G	A	C	T	T	G	A	T	T	C	C	A	A	G	C	A	A	G	C	A	G	C	T	A	C	C	G	G	C	A	A	G	C	A	T	G				
155	H155_62_A	ATCGSATCATCGCAGNAGAGAGCCGATGACGAAGAGCGG08TACCGAATCAAGCAGCG	A	C	G	S	A	T	C	A	T	C	G	C	A	N	A	G	A	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	C	G	C	G	C	G	T	A	C	C	G	A	T	C	A	G	C	A	C	C	G				
156	H156_62_A	ATCGSATCATCGCAGNAGAGAGCCGATGACGAAGAGCGCGCTACCGAATCAAGTACCG	A	C	G	S	A	T	C	A	T	C	G	C	A	N	A	G	A	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	C	A	A	G	C	A	G	C	G	T	A	C	C	G	A	T	C	A	G	T	A	C	C	G	
157	H157_62_A	ATCGSATCCHATTATNGAGAGCGCGCATGAGCAGCGG6GACTTGATCAAGTACCG	A	C	G	S	A	T	C	C	A	T	A	T	N	A	G	A	A	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	C	A	A	T	G	T	C	A	T	A	A	T	C	A	G	T	A	C	C	G					
158	H158_62_A	ATCGS6CTCATAGNAGAGAGAGCCGATGACGAAGAGCGG08TACCG66GCAAGCAGCG	A	C	G	S	G	C	T	C	A	T	A	G	N	A	G	A	A	A	A	C	G	A	C	C	A	T	G	A	C	C	G	A	A	G	C	A	A	T	G	A	C	C	G	C	G	T	A	C	C	G	G	C	A	A	G	C	A	C	C	G
159	H159_62_A	ATCTAJATCATATGNGTGTAGTGAATGATTCGAGAGAGCGGCTACCG66GCAAGCAGCG	A	C	T	A	L	A	T	C	C	A	T	A	T	T	G	N	G	A	T	A	G	T	G	A	A	C	T	T	G	A	T	T	C	C	G	A	A	G	C	G	C	G	T	A	C	C	G	G	C	A	A	G	C	A	C	C	G			
160	H160_62_A	ATCGSATCCHATTAGNAGAGAGCGGCTACCGAAGCAGCTCACTGATCAAGTGTTA	A	T	C	G	S	A	T	C	A	T	C	G	C	A	N	A	G	A	A	C	G	C	C	G	C	A	T	D	D	C	C	G	A	A	G	C	A	G	C	A	G	C	T	C	A	T	G	A	T	C	A	A	T	G	T	A				
161	H161_62_A	ATCGSATCATCGCAGNAGAGAGCCGATGACGAAGAGCGGCTACCGAATCAAGTACCG	G	T	C	G	S	A	T	C	A	T	C	G	C	A	N	A	G	A	A	A	C	G	C	C	G	C	A	T	G	A	C	C	G	A	A	G	C	G	C	G	C	T	A	C	C	G	A	T	C											

FIG. 17 CONT.



1073 Hap	I/U/G Hap
H1	H1
H2	H2
H3	H3
H4	H4
H5	H5
H6	H6
H7	H7
H8	H8
H9	H11
H10	H9
H11	H12
H12	H17
H13	H14
H14	H19
H15	H15
H16	H13
H17	H21
H18	H16
H19	H10
H20	H31
H21	H25
H22	H29
H23	H24
H24	H20
H25	H26
H26	H32
H27	H36
H28	H34
H29	H18

FIG. 19

H30	H41
H31	H30
H32	H28
H33	H27
H34	H43
H35	H22
H36	H39
H37	H53
H38	H48
H39	H23
H40	H37
H41	H45
H42	H23
H43	H5
H44	H50
H45	NP
H46	H2
H47	H35
H48	NP
H49	NP
H50	H10
H51	H3
H52	H6
H53	H51
H54	H12
H55	H33
H56	NP
H57	NP
H58	NP
H59	NP

FIG. 19 CONT.

H60	NP
H61	NP
H62	NP
H63	NP
H64	NP
H65	H49
H66	NP
H67	NP
H68	H24
H69	H56
H70	H37
H71	NP
H72	NP
H73	NP
H74	NP
H75	NP
H76	NP
H77	NP
H78	NP
H79	NP
H80	NP
H81	NP
H82	H42
H83	NP
H84	NP
H85	H47
H86	NP
H87	NP
H88	NP
H89	NP

FIG. 19 CONT.

H90	NP
H91	NP
H92	H5
H93	H9
H94	H1
H95	NP
H96	NP
H97	H54
H98	NP
H99	NP
H100	NP
H101	H40
H102	H14
H103	H3
H104	H2
H105	NP
H106	NP
H107	NP
H108	NP
H109	NP
H110	NP
H111	NP
H112	NP
H113	NP
H114	NP
H115	H32
H116	NP
H117	H28
H118	NP
H119	NP

FIG. 19 CONT.

H120	H2
H121	NP
H122	NP
H123	NP
H124	NP
H125	NP
H126	NP
H127	NP
H128	NP
H129	NP
H130	NP
H131	NP
H132	NP
H133	NP
H134	NP
H135	H12
H136	NP
H137	NP
H138	H21
H139	NP
H140	NP
H141	NP
H142	NP
H143	H34
H144	NP
H145	NP
H146	NP
H147	NP
H148	NP
H149	NP

FIG. 19 CONT.

H150	NP
H151	NP
H152	NP
H153	NP
H154	H9
H155	NP
H156	NP
H157	NP
H158	NP
H159	NP
H160	NP
H161	NP
H162	NP
H163	NP

FIG. 19 CONT.

Larger 1073 Hap	Incorporated 1073 Haps
H1	H94
H2	H46,H104,H120
H3	H51,H103
H5	H43,H92
H6	H52
H10	H93,H154
H11	H54,H135
H13	H102
H17	H138
H19	H50
H23	H68
H26	H115
H28	H143
H32	H117
H39	H42
H40	H70
H106	H150

FIG. 20

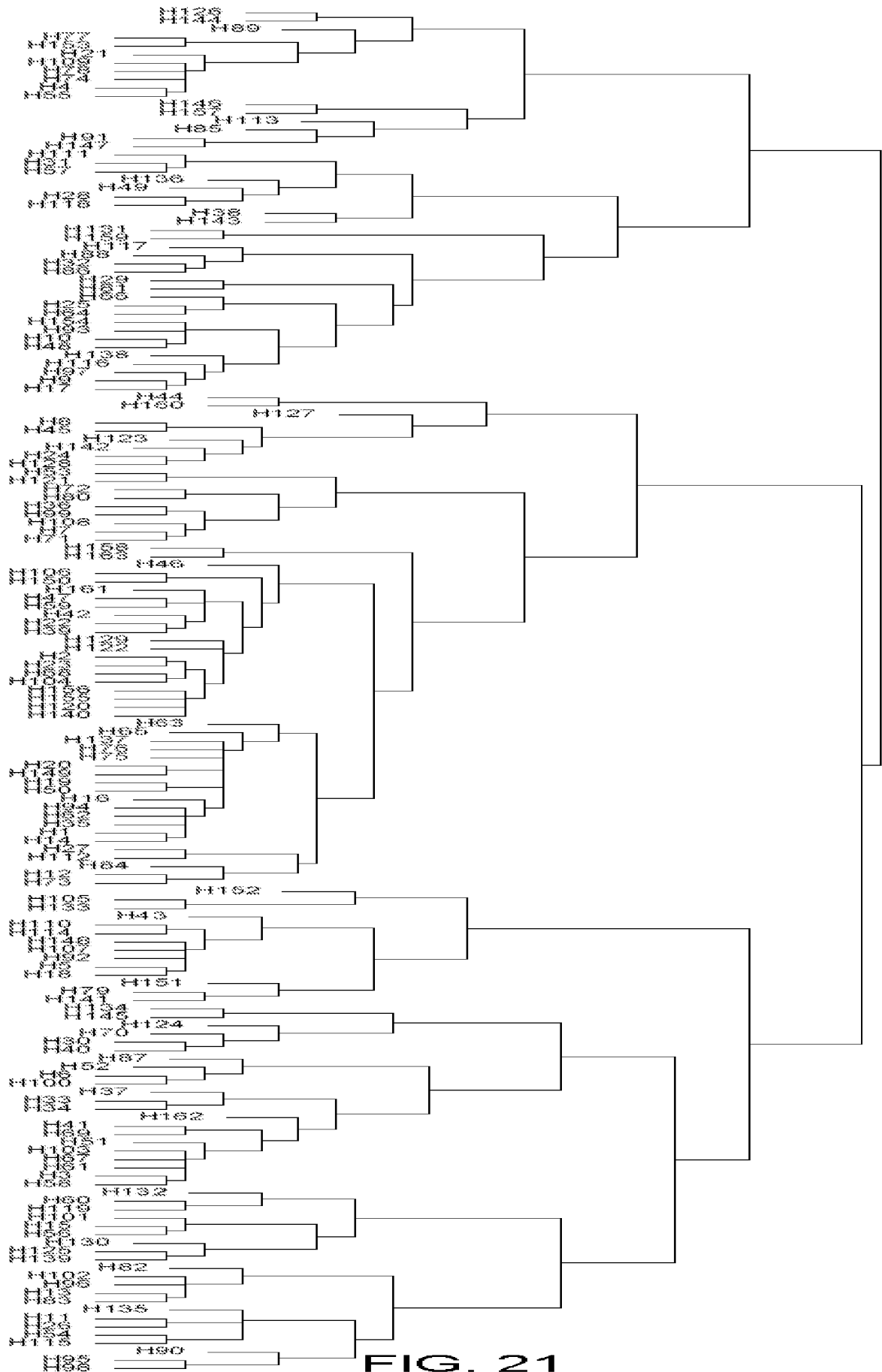


FIG. 21
68/154

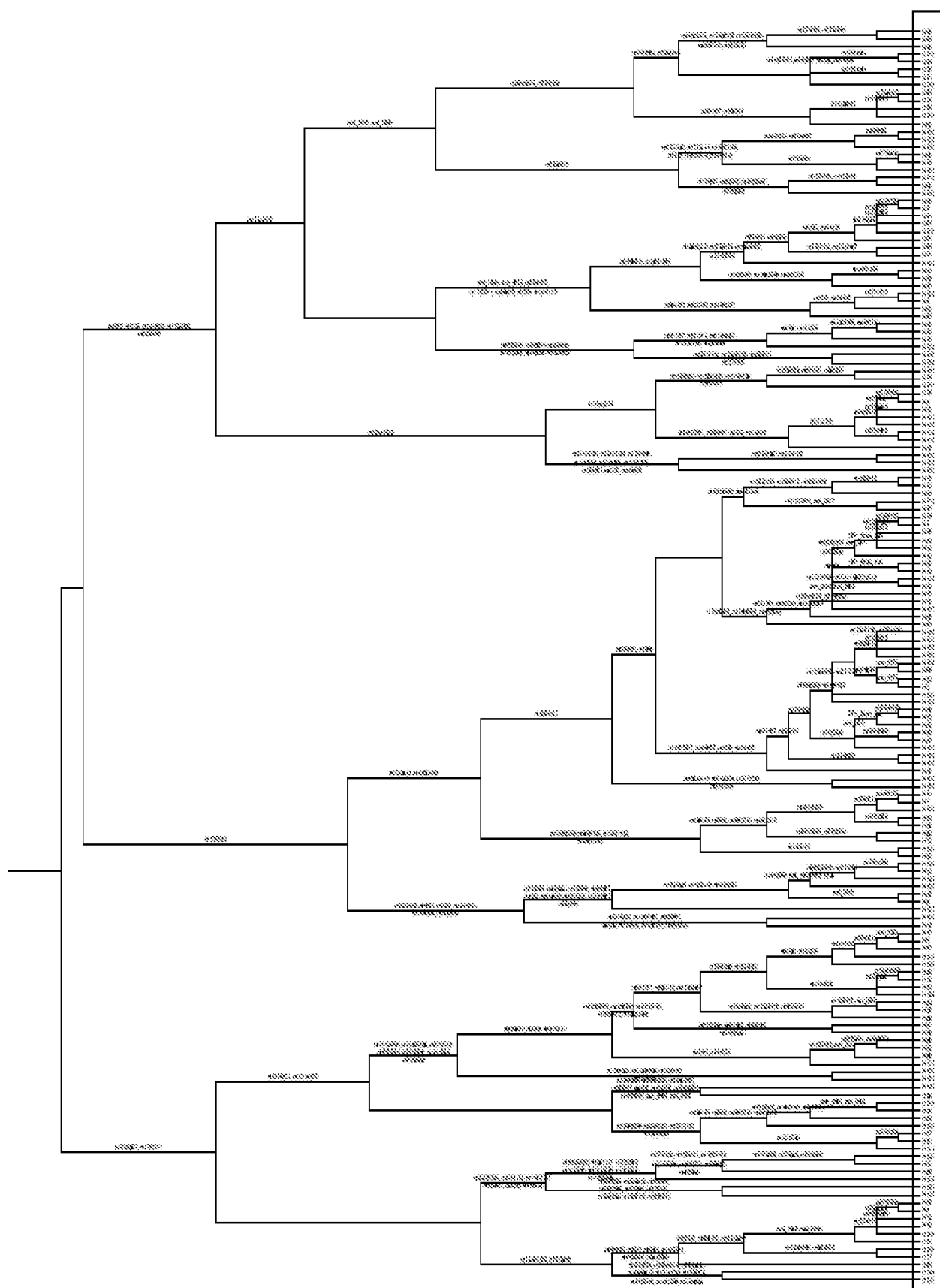


FIG. 21A

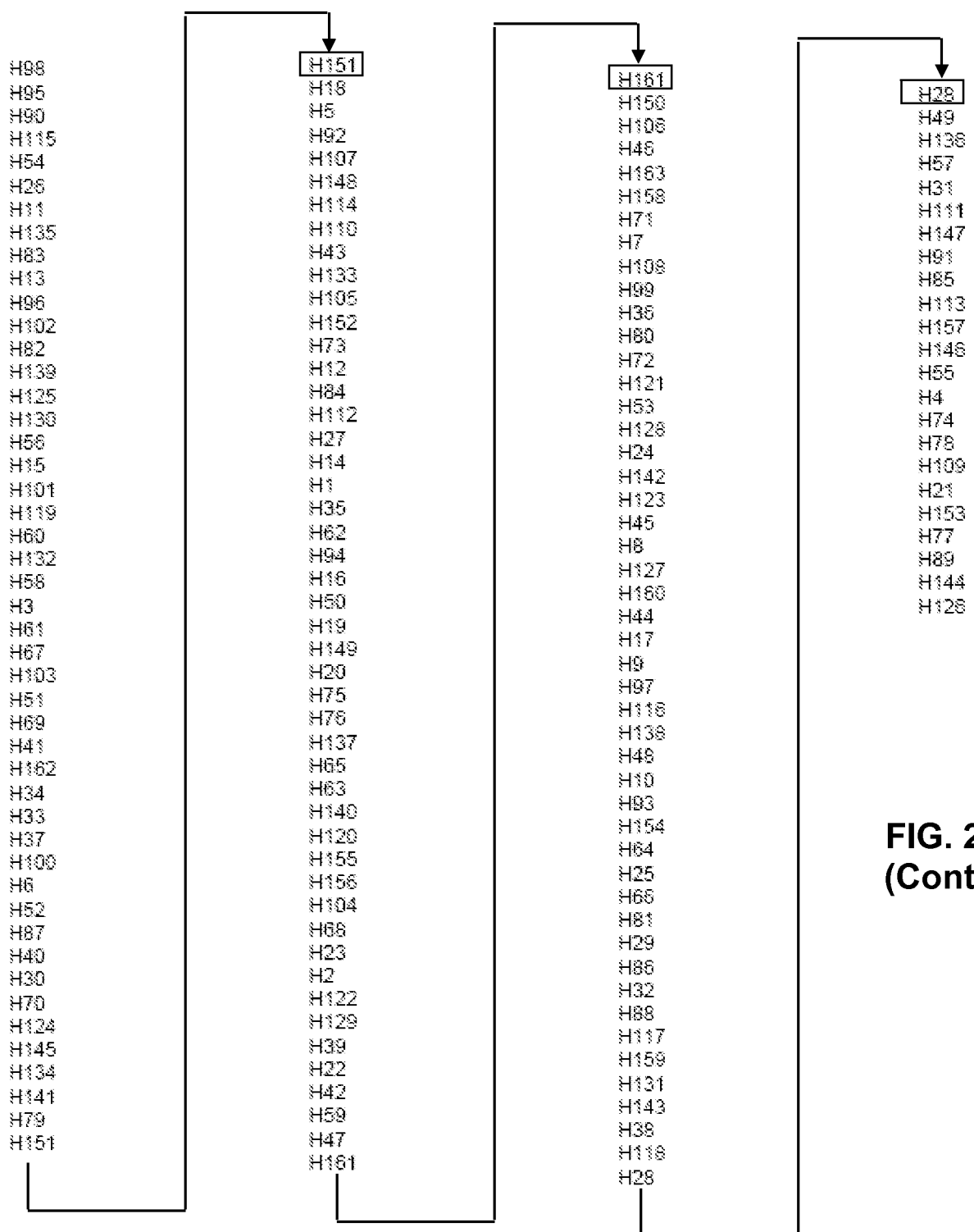


FIG. 21A
(Cont'd.)

Columns represent the boxed text from FIG. 21A
 Boxed text at the top of each column indicates a holdover from the previous column

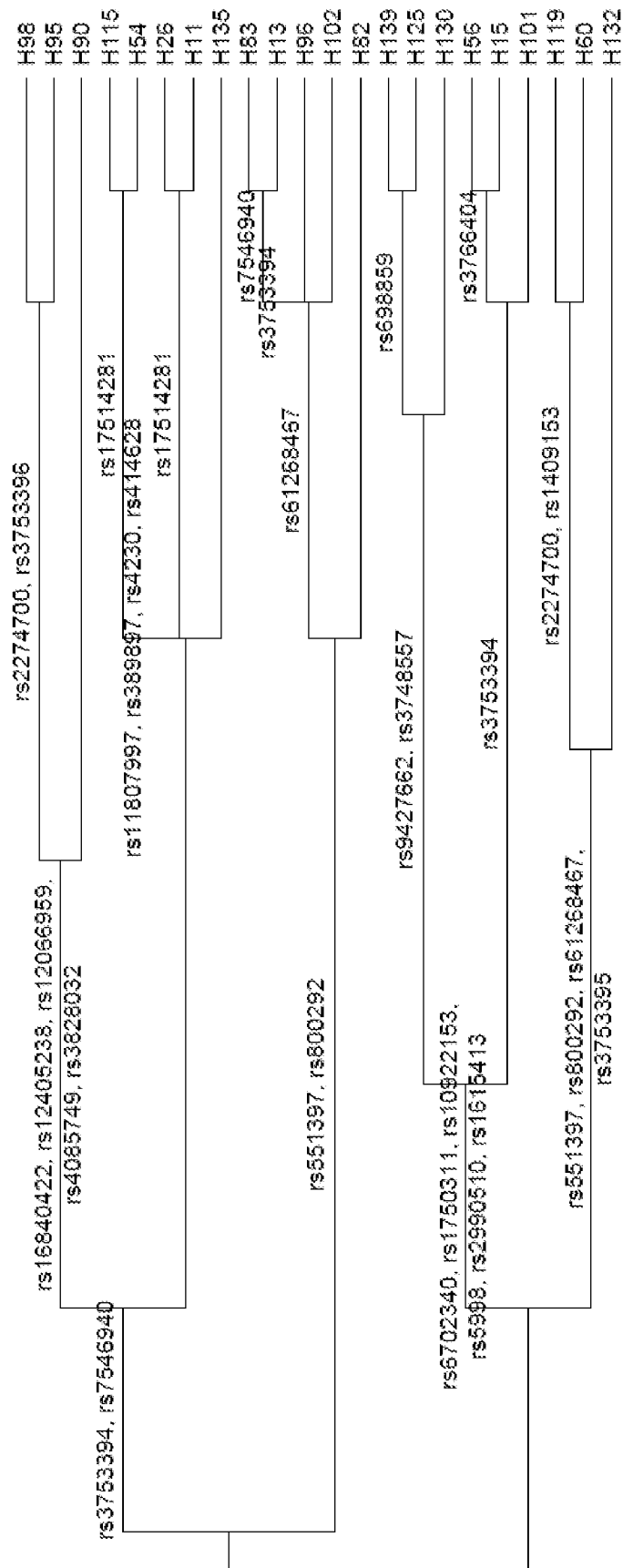


FIG. 21A-1

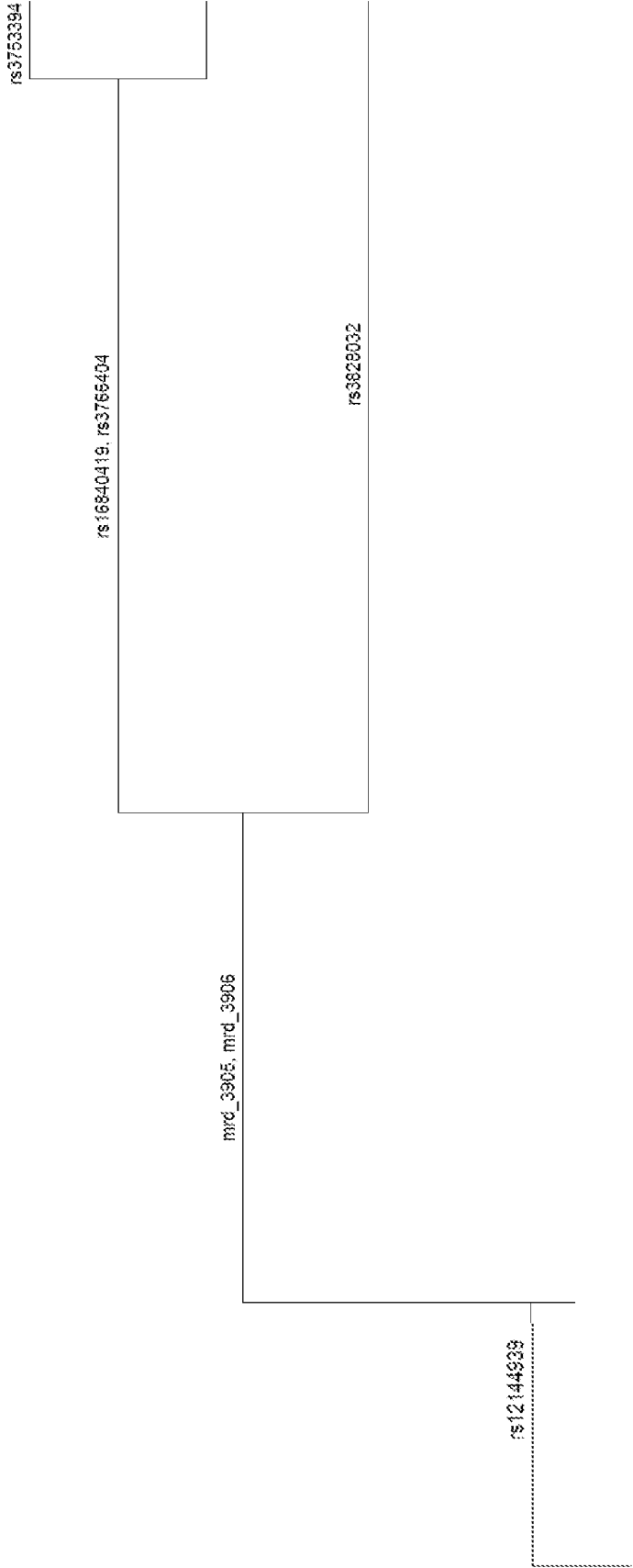


FIG. 21A-1 (cont'd.)

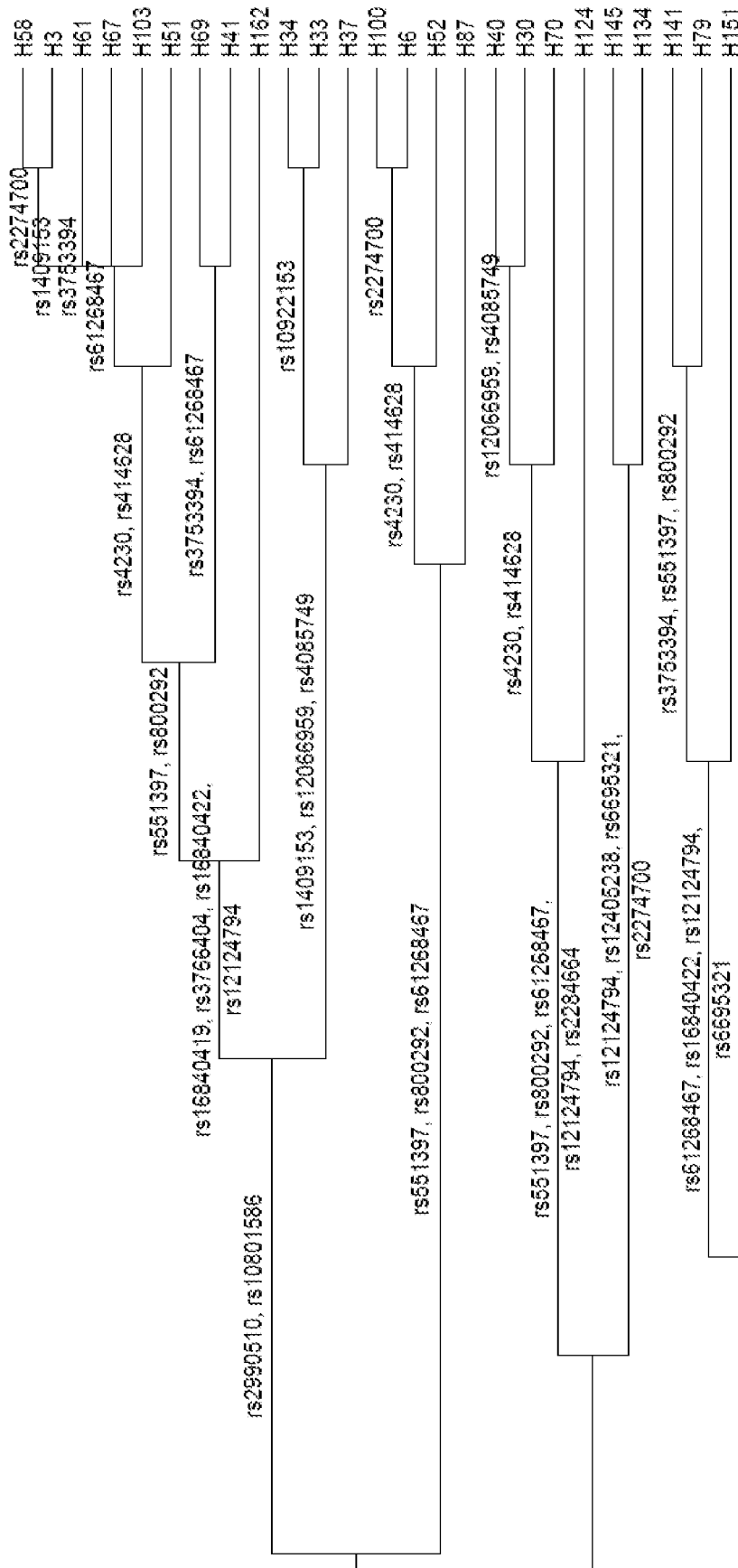


FIG. 21A-2

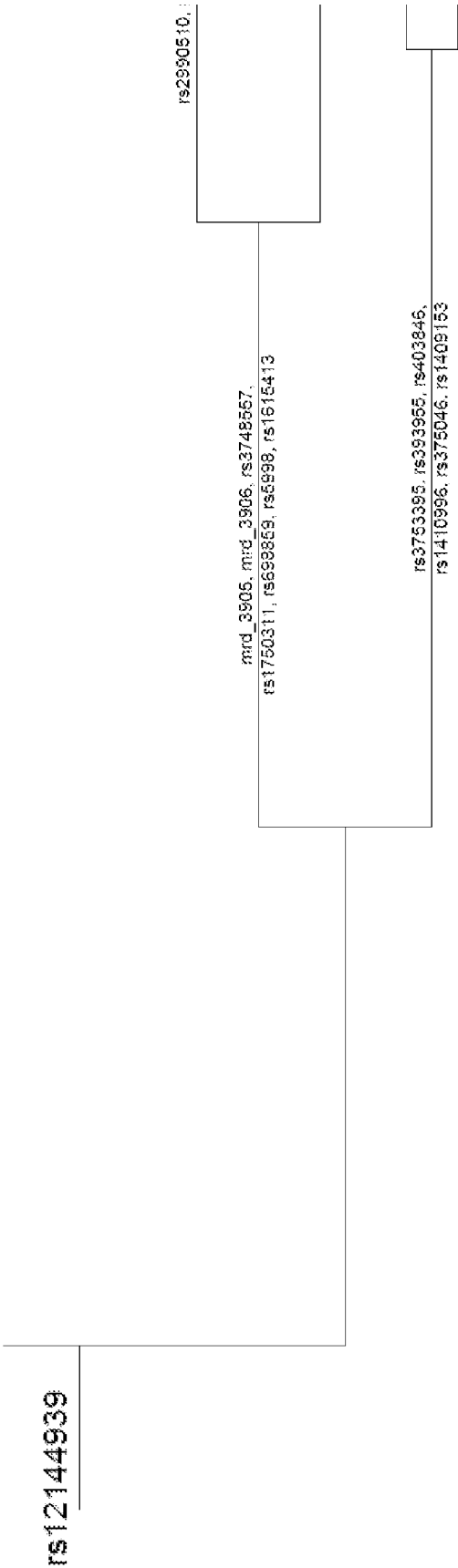


FIG. 21A-2 (cont'd.)

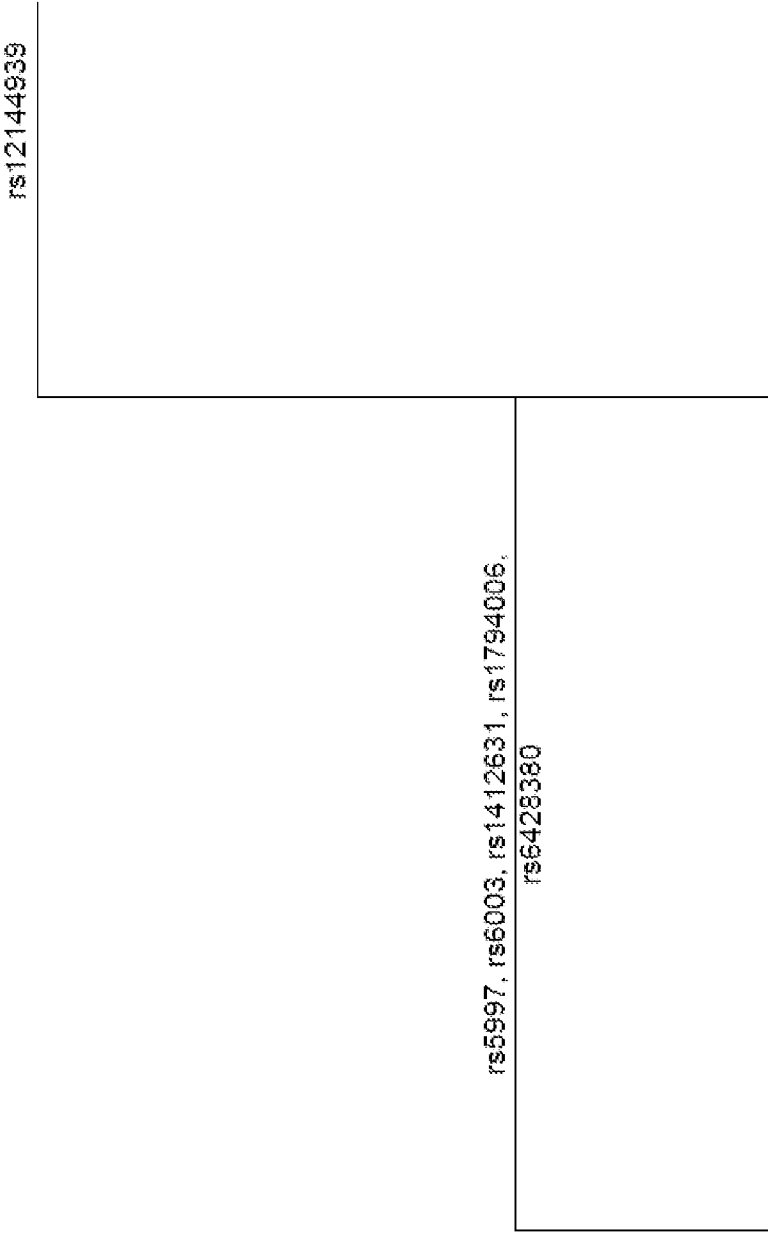


FIG. 21A-2 (cont'd.)

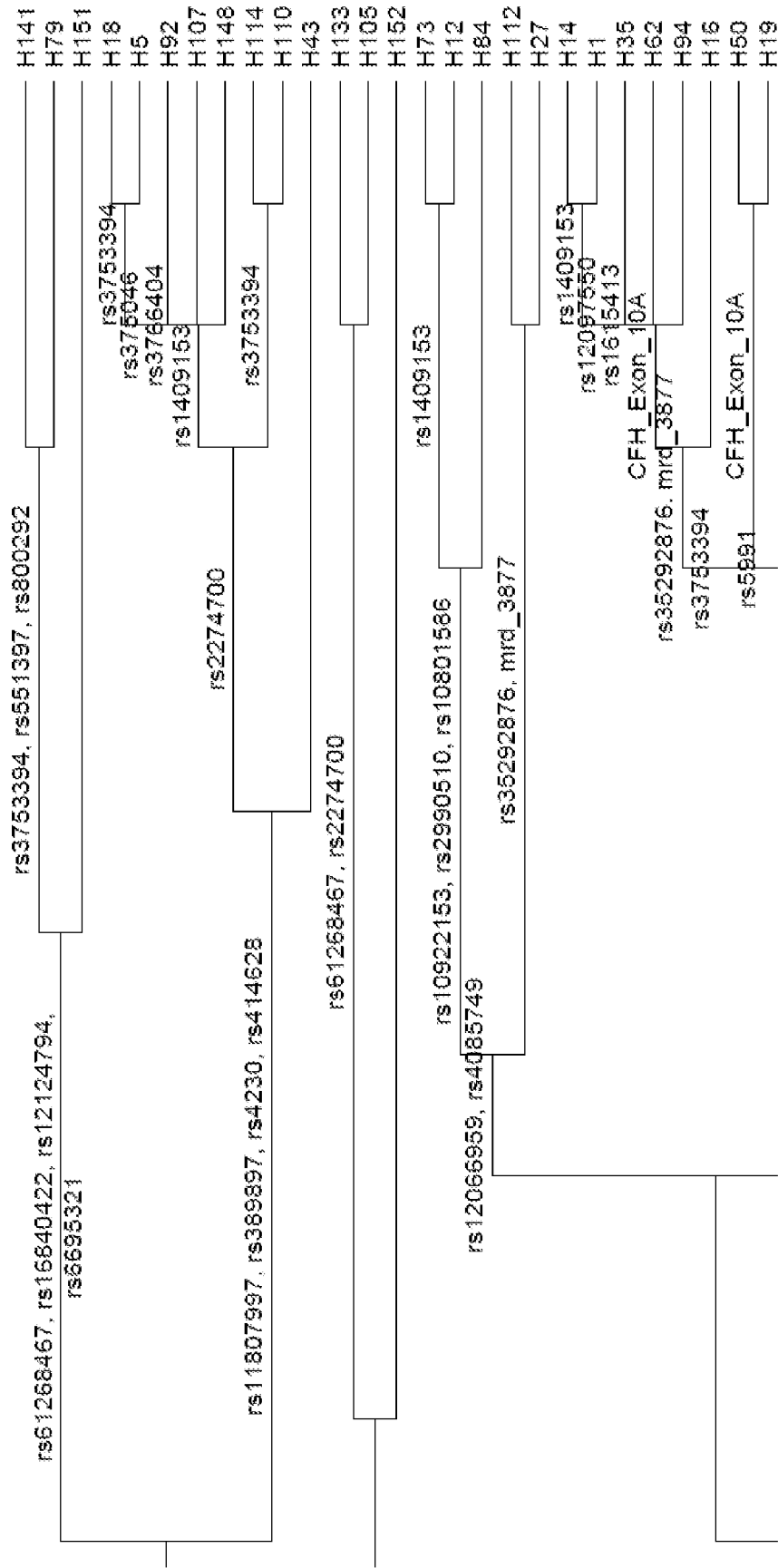


FIG. 21A-3

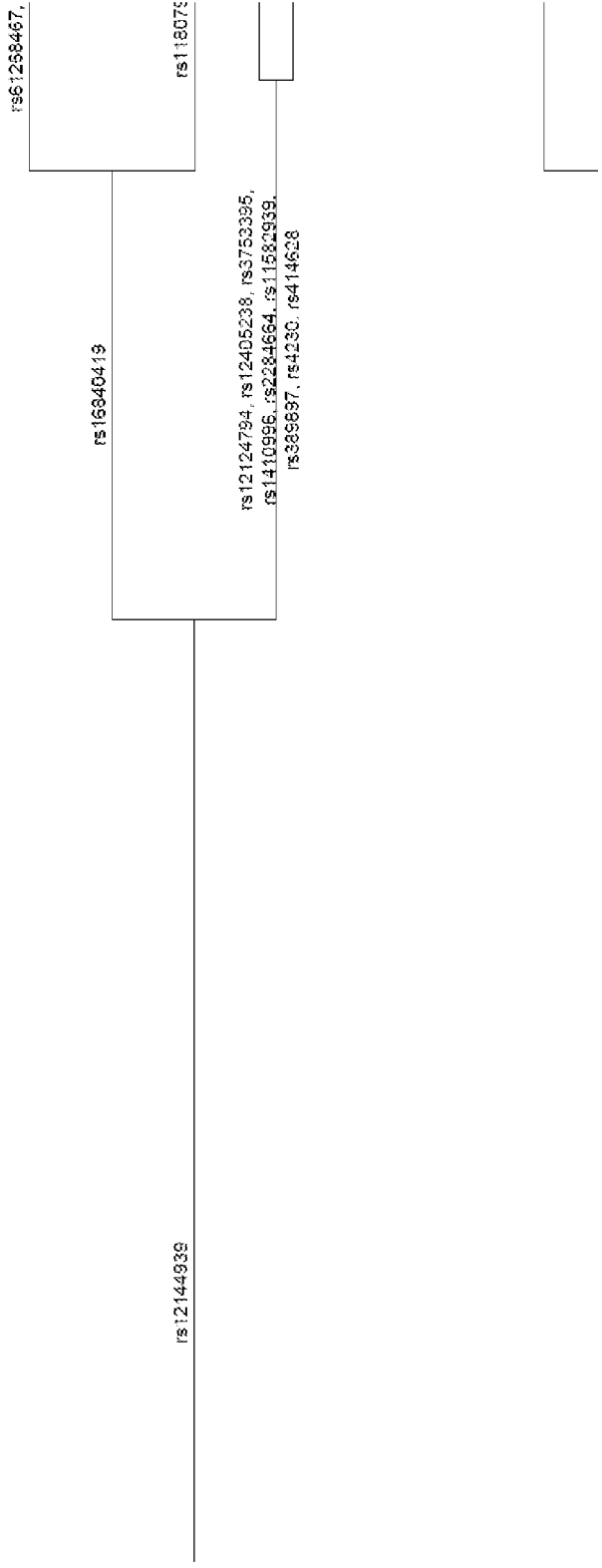


FIG. 21A-3 (cont'd.)

rs12144939

FIG. 21A-3 (cont'd.)

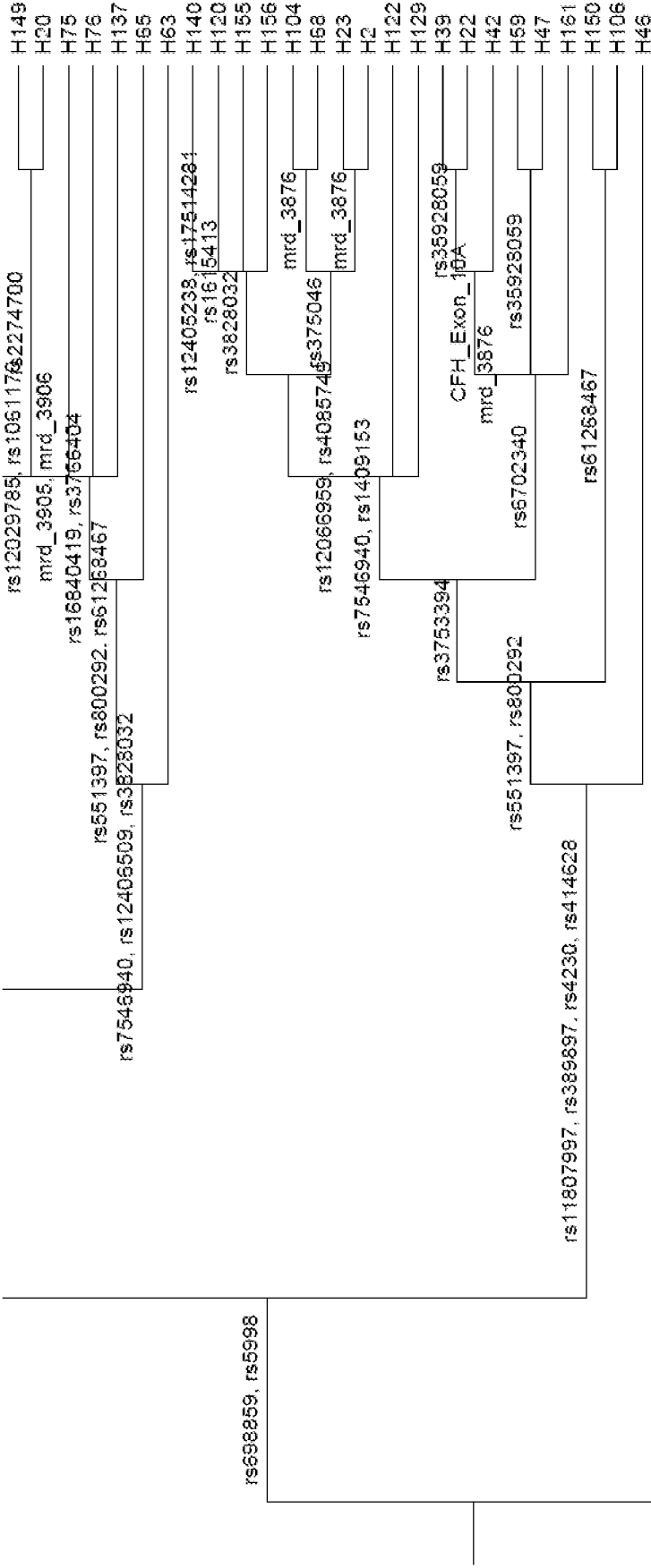


FIG. 21A-4

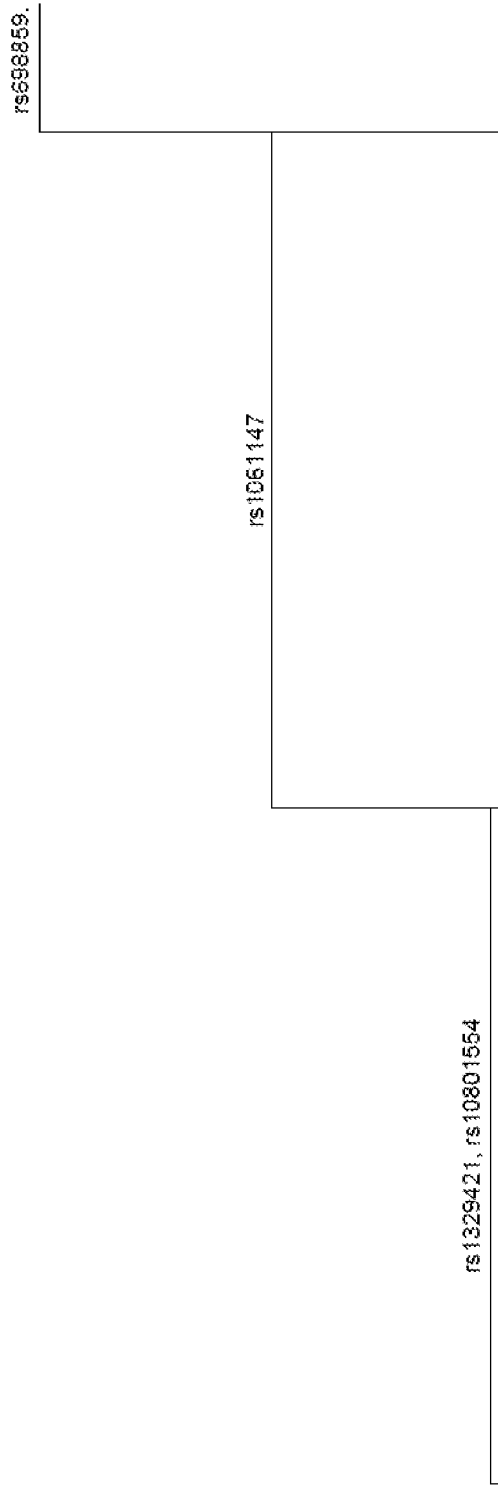


FIG. 21A-4 (cont'd.)

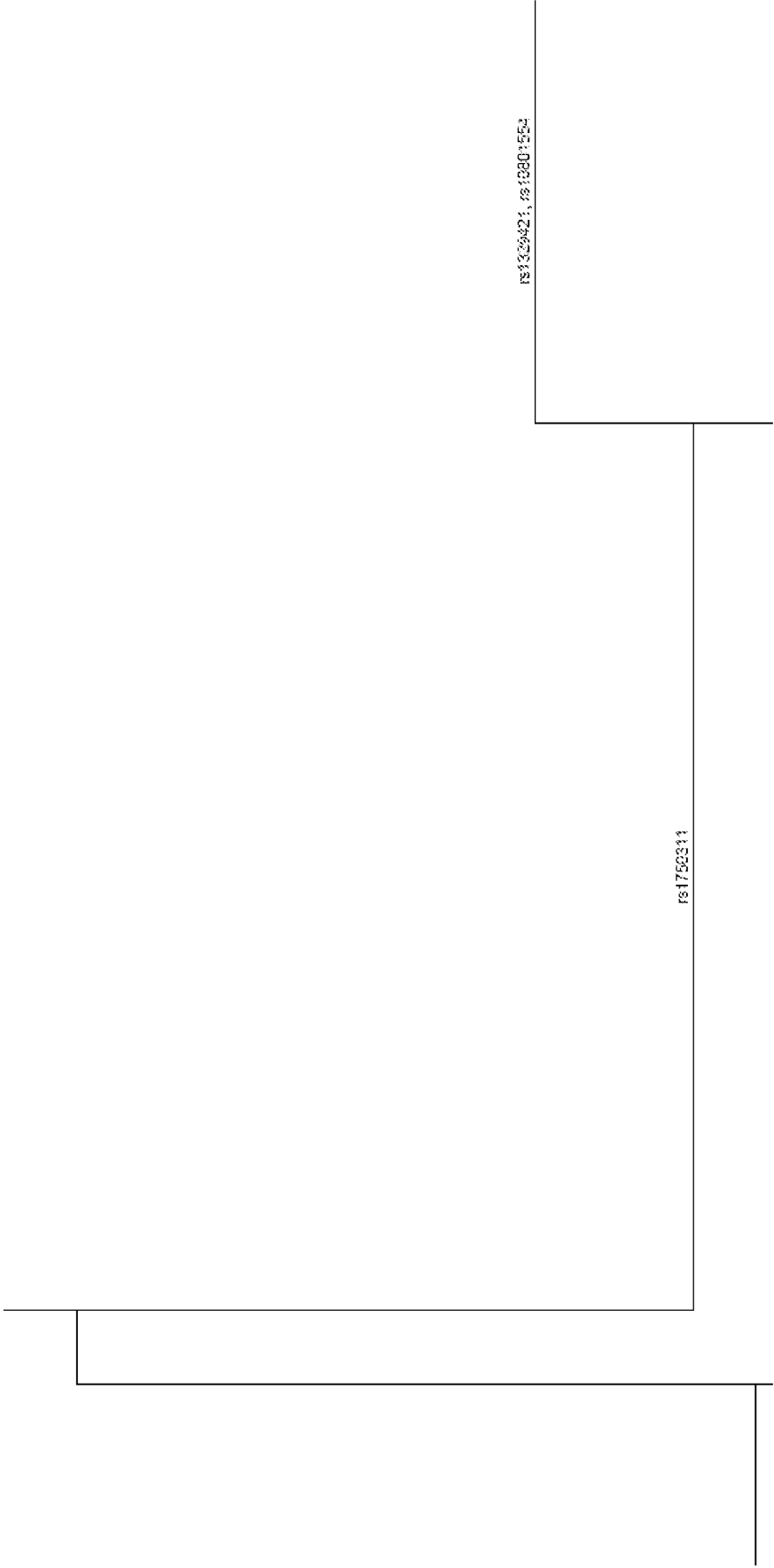


FIG. 21A-4 (cont'd.)

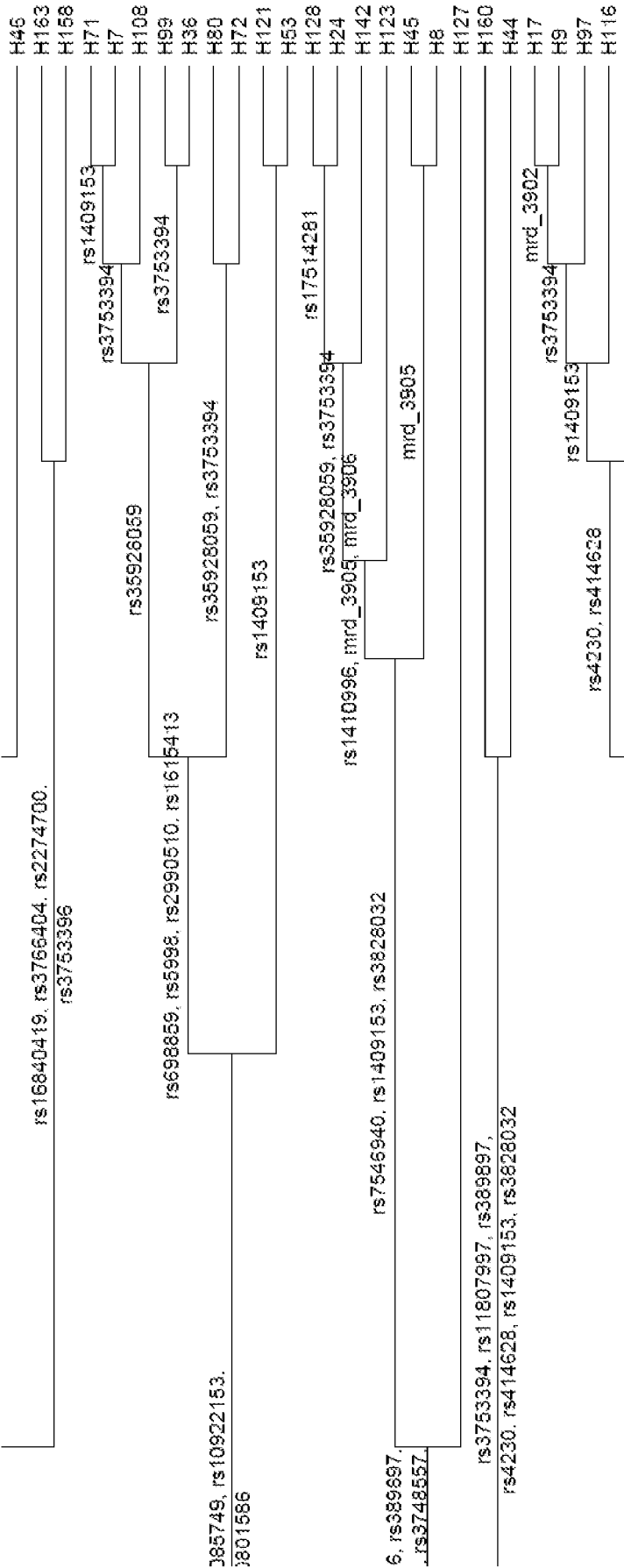


FIG. 21A-5

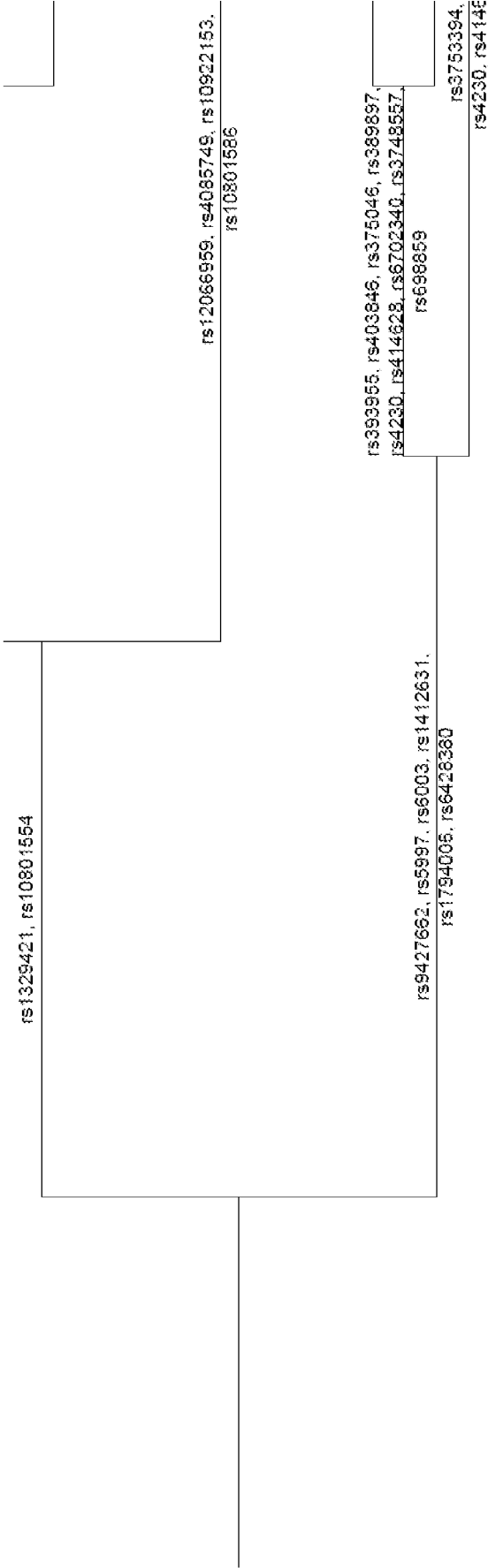


FIG. 21A-5 (cont'd.)

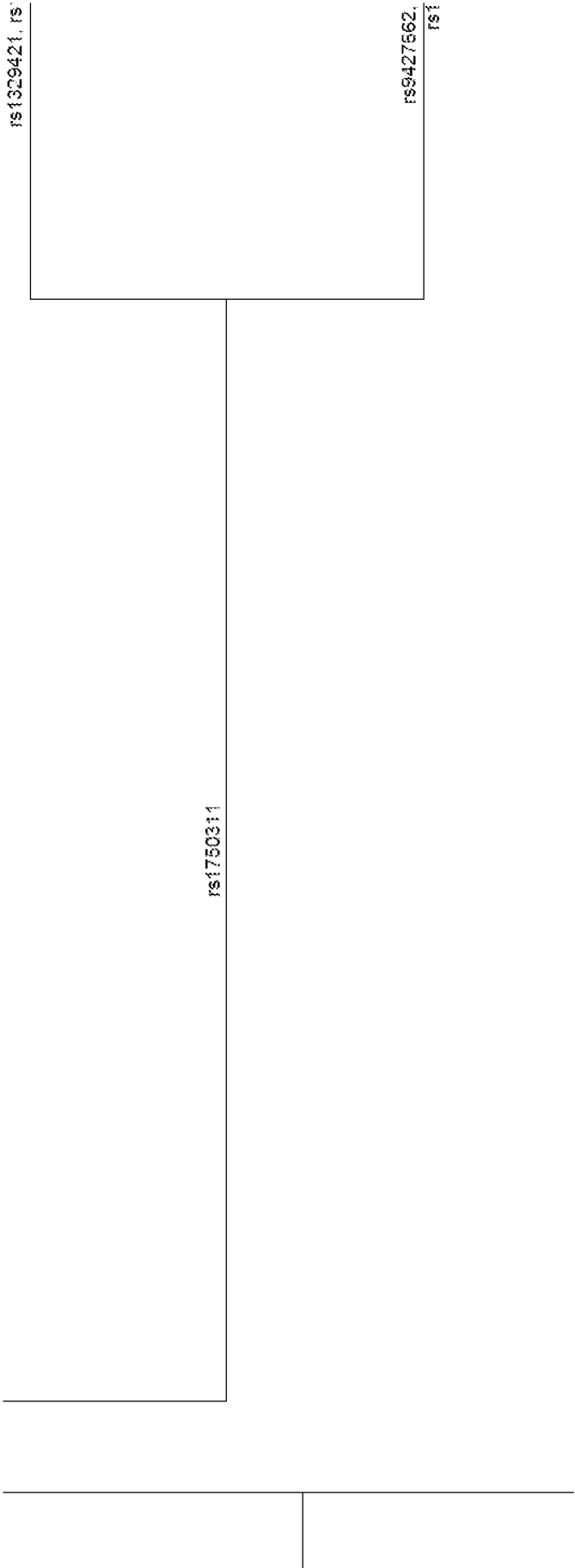


FIG. 21A-5 (cont'd.)

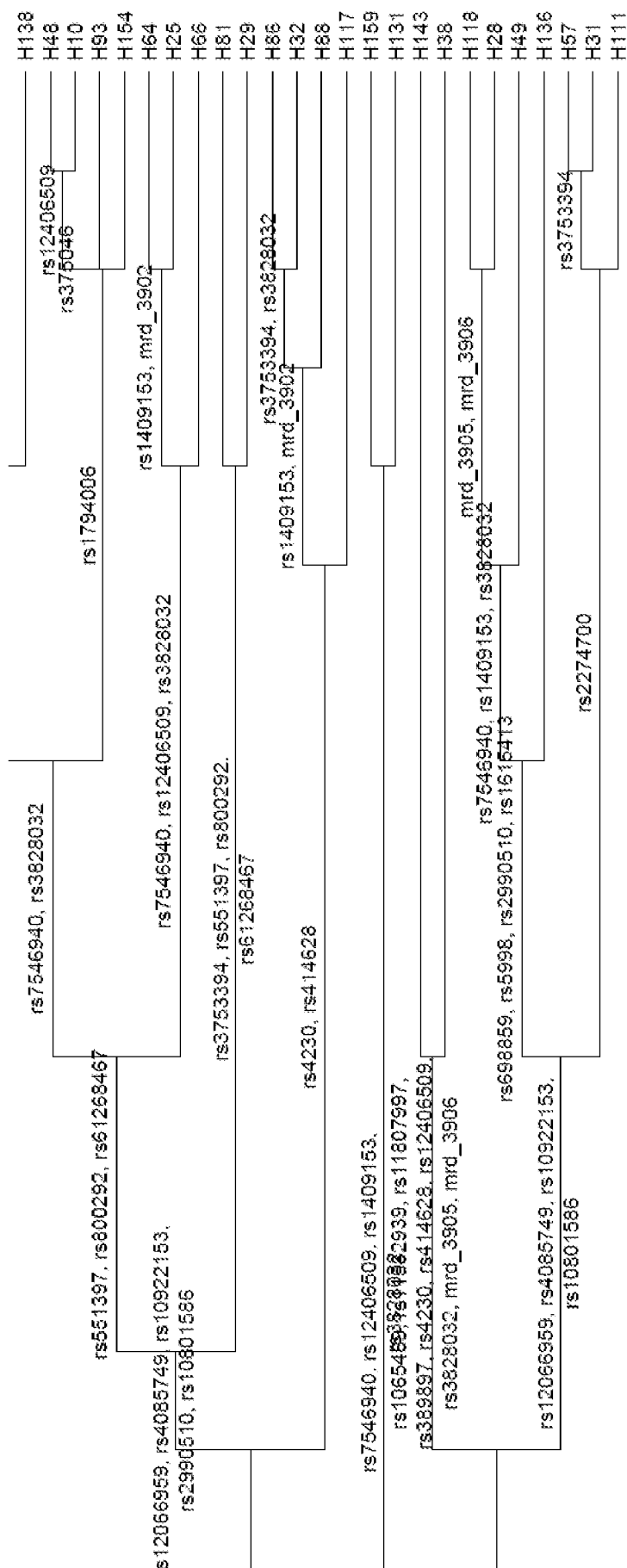


FIG. 21A-6

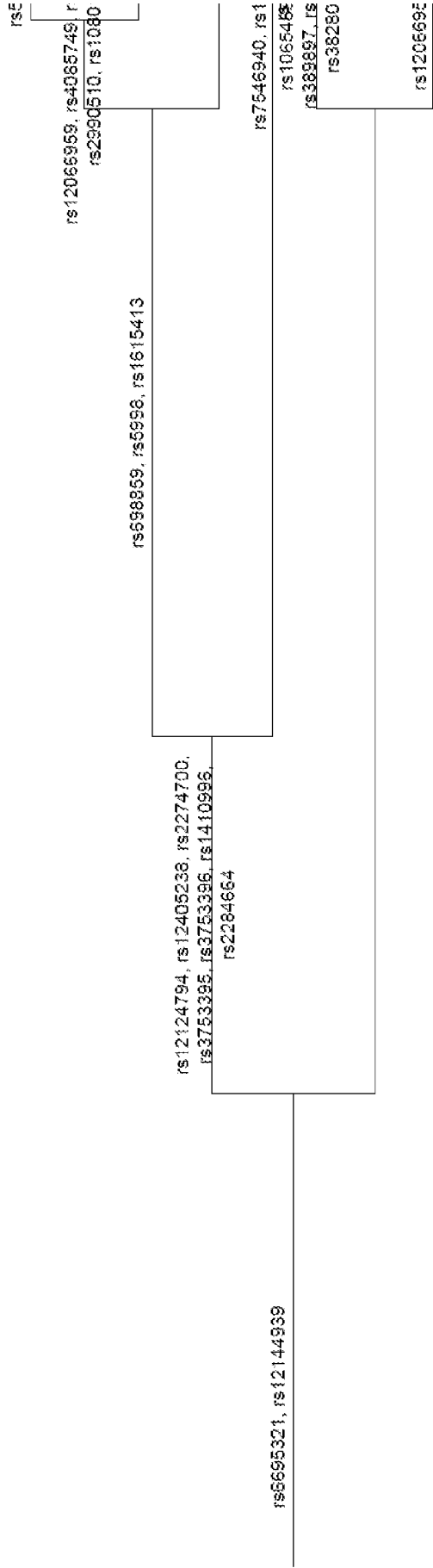


FIG. 21A-6 (cont'd.)

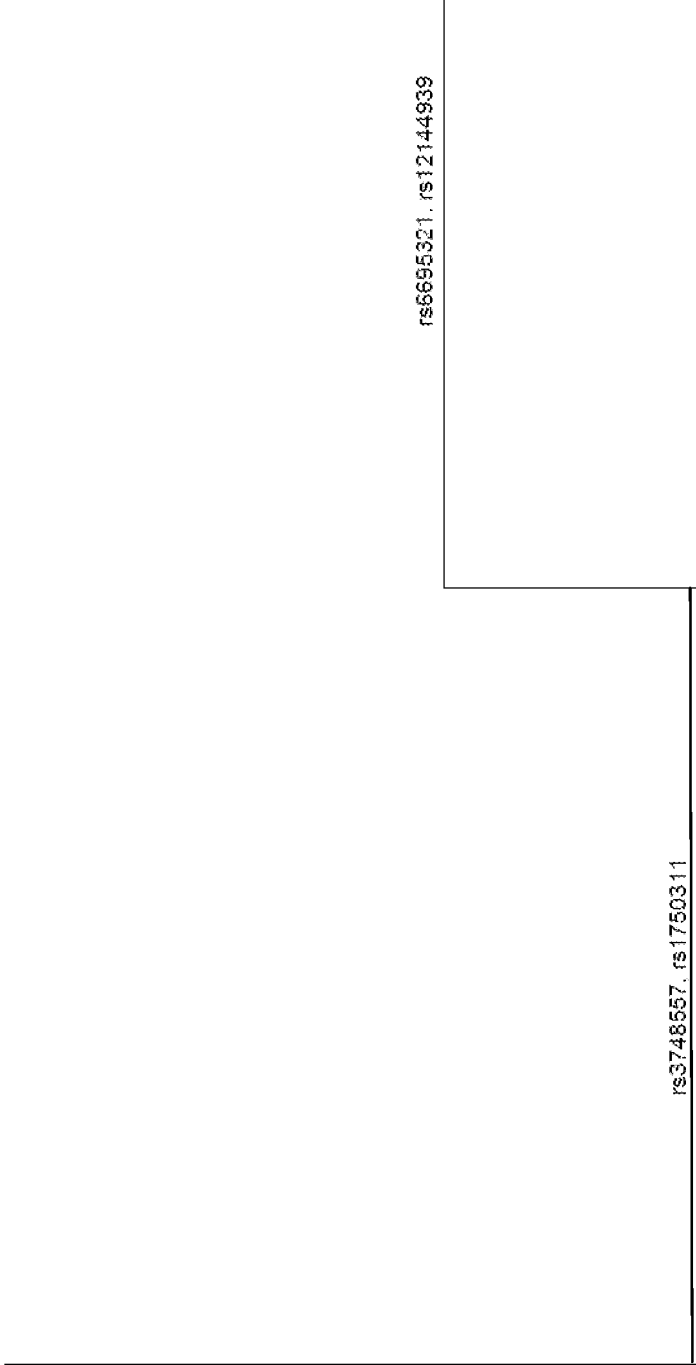
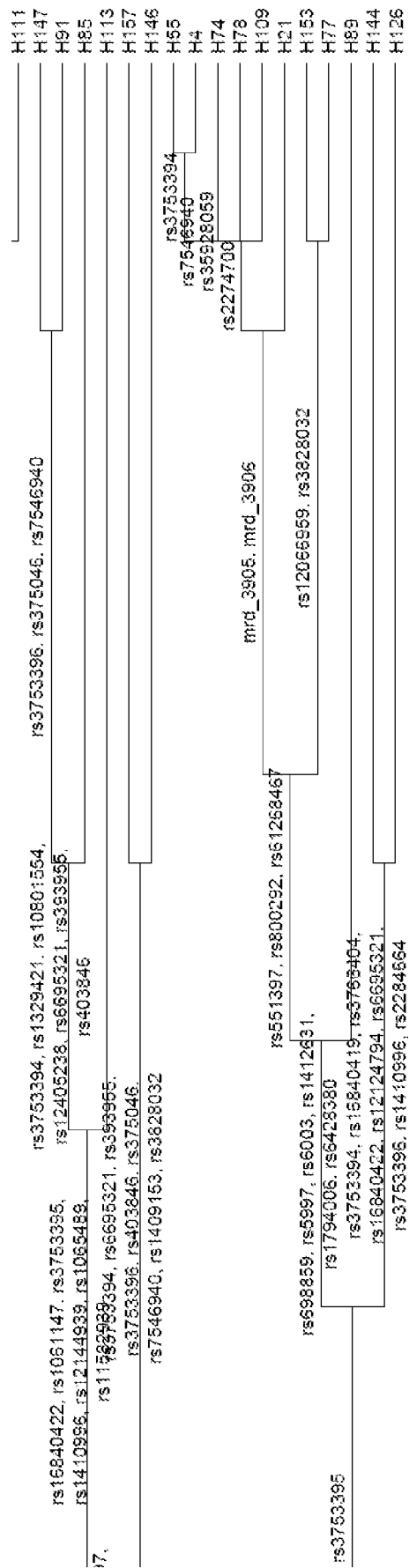


FIG. 21A-6 (cont'd.)



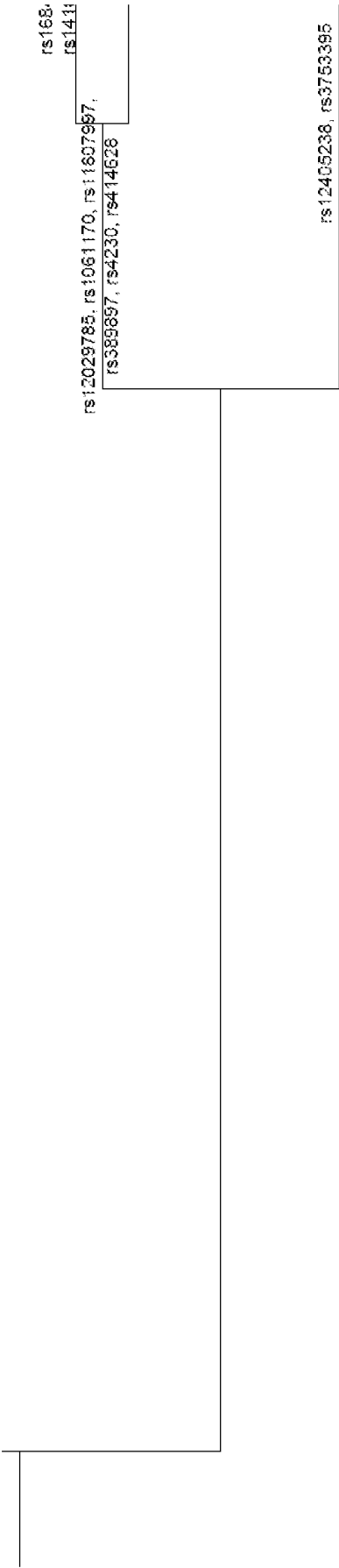


FIG. 21A-7 (cont'd.)

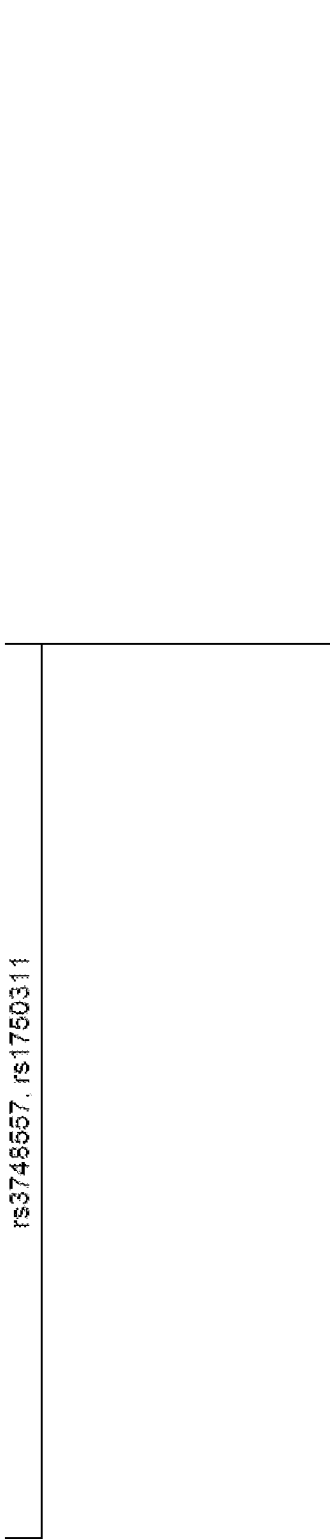


FIG. 21A-7 (cont'd.)

Q38	ID	Haplotype	Haplotype Sequence	r35928059	r3593394	r551397	r36840419	r3766404	r36840422	r3329421	r310801554	r31061170	r36695321	r32274700	r3753895	r3939555	r3753996	r403846	r3212144939	r32284664	r35292876	r31065489	r311562939	mrd_3876 (r3112797613)	mrd_3877	r3546940	r32406509 (mrd_3878)	r3409153	r32066959	mrd_3902 (r39351096)	r3828032	mrd_3905 (r35816959)	mrd_3906 (r374188363)	r3427662	r3748557	r32097550	r310922153	r3598	r32990510	r310801586	r35991	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r31750311	r317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FIG. 22

196	H33_51_A	ADCGATCCTTTATGACAAGGCGCGGACGCGGGTGGGTATCTTCATCC	A	C	C	G	A	T	C	C	T	T	A	T	G	A	C	A	A	G	G	C	C	G	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	C	T	C	A	T	C	C		
197	H34_51_A	ATCGATCCTTTATATGAATCGCGGATACGGGGTTCCTGGTTTGGTCC	A	T	C	G	A	T	C	C	T	T	A	T	A	G	A	T	C	C	G	C	C	G	G	A	T	A	C	G	C	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C	C			
198	H35_51_A	ACTAATCCTTTTCATAAGAGTGGCGGATACGGGGTTCCTGGGCTTGCTCC	A	C	T	A	A	T	C	C	T	T	T	C	A	T	A	G	A	G	T	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	T	C	G	G	C	T	C	G	T	C		
199	H36_51_A	ACTAATCCTTTTCATAAGAGTGGCGGATGCGGGGTTCCTGGGCTTGCTCC	A	C	T	A	A	T	C	C	T	T	T	C	A	T	A	G	A	G	T	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	G	T	A	T	C	T	C	A	T	C		
200	H37_51_A	ATCGATCCTTTACGAAGGGGCTTCGGATACGGGGTTCCTGGTTTGGTCC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C			
201	H38_51_A	ACCGATCAACATGACAAGGCGCGGACGCGGGGTACGGGGCTTGCTCC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G	G	A	C	G	C	C	G	G	T	T	A	C	G	C	G	G	C	T	C	G	T	C			
202	H39_51_A	ACCGATCCTTTTCATAAGAGTGGCGGATACGGGGTTCCTGGTTTGGTCC	A	C	C	G	A	T	C	C	T	T	T	C	A	T	A	G	A	G	T	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C			
203	H40_51_A	ACTAATCCTTTTCATAAGAGTGGCGGACGCGGGGTTCCTGGTTTGGTCC	A	C	T	A	A	T	C	C	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C	
204	H41_51_A	ACCGATCAACATGACAAGGCGCGGACGCGGGGTACGGGGCTTGCTCC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G	G	A	C	G	C	C	G	G	T	T	A	C	G	C	G	G	C	T	C	A	T	C			
205	H42_51_A	GTGGATCAACATGACAAGGCGCGGAGCGGGGTTCGTTTGGTCC	G	T	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	T	C	G	T	T	C	A	T	C			
206	H43_51_A	GTGGATCCTTTATGACAAGGCGCGGATACGGGGTTCCTGGTTTGGTCC	G	T	C	G	A	T	C	C	T	T	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C		
207	H44_51_A	ATCGATCCTTTATATGAATCGCGGACGCGGGGTTCCTGGTTTGGTCC	A	T	C	G	A	T	C	C	T	T	A	T	A	G	A	T	C	C	G	C	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C	
208	H45_51_A	ATCGATCCTTTATATGAATCGGAGTTCGAGTTCGGGGTTCCTGGTTTGGTCC	A	T	C	G	A	T	C	C	T	T	A	T	A	G	A	T	C	C	T	T	C	G	A	G	T	G	C	G	G	T	T	G	C	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C
209	H46_51_A	ACCGATCCTTTACGAAGGGGCTTCGAATGCAAGTAGATTCTTCATCC	A	C	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	A	A	T	G	C	A	A	A	G	T	A	G	A	T	A	T	C	T	C	A	T	C	C		
210	H47_51_A	ACCGATCAACATGACAAGGCGCGGACGCGGGGTCTGATCATCTTACTT	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G	G	A	C	G	C	G	G	T	C	T	G	A	T	C	A	T	C	T	T	A	C	T	T		
211	H48_51_A	ACCGGTTCTTTATGAAGATCGCGGATGCAAGGGTTCCTGGTTTGGTCC	A	C	C	G	T	C	C	T	T	A	T	A	G	A	T	C	C	G	C	C	G	G	A	T	C	C	G	G	A	T	G	C	A	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C
212	H49_51_A	ACTAATCCTTTACGAAGGGGCTTCGAGTGCAGGGTTCCTGGTTTGGTCC	A	C	T	A	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	A	G	T	G	C	A	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C	C		
213	H50_51_A	ATCGATCCTTTACGAAGGGGCTTCGAATGCAAGGGTTCCTGGTTTGGTCC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	A	A	T	G	C	A	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C	C		
214	H51_51_A	GTGGATCCTTTATGACAAGGCGCGGATGCGGGGTTCGTTTGGTCC	G	T	C	G	A	T	C	C	T	T	A	T	G	A	C	A	A	G	C	C	C	C	G	A	T	C	C	G	G	T	T	G	C	G	G	T	T	G	C	G	T	A	T	C	T	C	A	T	C
215	H52_51_A	ADCGATCCTTTATGACAAGGCGCGGACGCGGGGTTCCTGGTTTGGTCC	A	C	C	G	A	T	C	C	T	T	A	T	G	A	C	A	A	G	C	C	C	C	G	A	T	G	C	C	C	C	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C	C		
216	H53_51_A	ACCGATCCTTTATATGAAGATCGCGGATACGGGGTTCCTGGTTTGGTCC	A	C	C	G	A	T	C	C	T	T	A	T	A	G	A	T	C	C	G	C	C	C	G	G	A	T	A	C	G	C	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C	C		
217	H54_51_A	ACTAATCCTTTACGAAGGGGCTTCGAGTGCAGGGTTCCTGGTTTGGTCC	A	C	T	A	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	A	A	T	G	C	A	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C	C		
218	H55_51_A	ACCGATCCTTTACGAAGGGGCTTCGGATGTGGGGTTCGTTTGGTCC	A	C	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	G	A	T	G	T	G	G	T	T	G	C	G	C	G	G	T	T	G	C	T	C	A	T	C	
219	H56_51_A	ACTAATCCTTTTCATAAGAGTGGCGGACGCGGGGTTCCTGGTTTGGTCC	A	C	T	A	A	T	C	C	T	T	T	C	A	T	A	G	A	G	T	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C	C		
220	H57_51_A	ACCGATCCTTTTCATAAGAGTGGCGGATACGGGGTTCCTGGTTTGGTCC	A	C	C	G	A	T	C	C	T	T	T	C	A	T	A	G	A	G	T	C	G	C	C	G	G	A	T	A	C	G	C	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C		
221	H58_51_A	ATCGATCAACATGACAAGGCTTCGAATGCAAGTAGATTCTTCATCC	A	T	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	G	C	C	T	T	C	G	A	A	T	G	C	A	A	A	G	T	T	A	G	A	T	A	T	C	T	C	A	T	C		
222	H59_51_A	ACCGATCCTTTACGAAGGGGCTTCGAATGCAAGTAGATTCTTACTT	A	C	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	A	A	T	G	C	A	A	A	G	T	T	A	G	A	T	A	T	C	T	T	A	C	T	T	
223	H60_51_A	ACCGATCAACATGACAAGGCGCGGACGCGGGGTTCCTGGTTTGGTCC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G	G	A	T	A	C	G	C	C	G	G	T	T	G	C	G	C	G	G	C	T	C	G	T	C	
224	H61_51_A	ACTAGCTCTTTATGAAGATCGCGGATGCAAGTAGATTCTTCATCC	A	C	T	A	G	C	T	C	T	T	A	T	A	G	A	T	C	C	G	C	C	G	A	A	T	G	C	A	A	A	G	T	T	A	G	A	T	A	T	A	T	C	T	C	A	T	C		
225	H62_51_A	ACTAATCCTTTTCGAAGAGTGGCGGATACGGGGTTCCTGGTTTGGTCC	A	C	T	A	A	T	C	C	T	T	T	C	G	T	A	A	G	A	G	T	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C		
226	H63_51_A	ATCGATCCTTTTCATAAGAGTGGCGGATACGGGGTTCCTGGTTTGGTCC	A	T	C	G	A	T	C	C	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G	G	A	T	A	C	G	C	G	G	T	T	G	C	T	C	G	T	T	C	G	T	C		
227	H64_51_A	ACTAATCCTTTACGAAGGGGCTTCGAGTACGGGGTTCCTGGTTTGGTCC	A	C	T	A	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	T	C	G	G	A	T	A	C	G	G	A	T	A	C	G	G	T	T	G	C	T	C	G	T	C	C		
228	H65_51_A	ATCGATCAACATGACAAGGCGCGGAGCGGGGTTCCTGGTTTGGTCC	A	T	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G	G	A	T	G	C	G	C	G	G	T	T	G	C	G	C	G	G	C	T	C	A	T	C	C	

FIG. 22 CONT.

Haplotype	Freq.	Case, Control Ratio Counts	Case,Control Freqs	Chi Square	P Value
H1_51_A	0.205	250.5 : 877.4, 118.8 : 500.6	0.222, 0.192	2.205	0.1375
H2_51_A	0.167	238.3 : 889.6, 63.1 : 556.3	0.211, 0.102	33.51	7.10E-09
H3_51_A	0.078	68.7 : 1059.2, 71.6 : 547.8	0.061, 0.116	16.21	5.67E-05
H4_51_A	0.063	80.0 : 1048.0, 34.0 : 585.4	0.071, 0.055	1.698	0.1925
H5_51_A	0.058	48.9 : 1079.0, 56.0 : 563.4	0.043, 0.090	15.61	7.78E-05
H6_51_A	0.038	40.0 : 1088.0, 29.3 : 590.1	0.035, 0.047	1.474	0.2246
H7_51_A	0.034	37.8 : 1090.2, 22.8 : 596.6	0.034, 0.037	0.13	0.7185
H8_51_A	0.024	27.5 : 1100.4, 15.0 : 604.4	0.024, 0.024	0.001	0.9718
H9_51_A	0.019	24.3 : 1103.6, 10.0 : 609.4	0.022, 0.016	0.627	0.4284
H10_51_A	0.019	11.0 : 1117.0, 23.0 : 596.4	0.010, 0.037	15.77	7.14E-05
H11_51_A	0.017	21.9 : 1106.0, 9.0 : 610.4	0.019, 0.015	0.554	0.4568
H12_51_A	0.015	18.5 : 1109.5, 9.1 : 610.3	0.016, 0.015	0.068	0.7938
H13_51_A	0.012	14.3 : 1113.6, 7.8 : 611.6	0.013, 0.013	0	0.9848
H14_51_A	0.012	8.4 : 1119.5, 12.6 : 606.8	0.007, 0.020	5.559	0.0184
H15_51_A	0.012	14.6 : 1113.4, 6.4 : 613.0	0.013, 0.010	0.217	0.6411
H16_51_A	0.01	15.9 : 1112.1, 2.9 : 616.5	0.014, 0.005	3.282	0.0701
H17_51_A	0.01	12.1 : 1115.9, 6.0 : 613.4	0.011, 0.010	0.043	0.8358
H18_51_A	0.01	14.3 : 1113.7, 3.0 : 616.4	0.013, 0.005	2.485	0.115
H19_51_A	0.01	11.0 : 1117.0, 6.1 : 613.3	0.010, 0.010	0.002	0.9664
H20_51_A	0.009	9.5 : 1118.5, 6.1 : 613.3	0.008, 0.010	0.088	0.7671
H21_51_A	0.008	9.0 : 1118.9, 5.0 : 614.4	0.008, 0.008	0	0.9866
H22_51_A	0.008	6.0 : 1122.0, 8.0 : 611.4	0.005, 0.013	2.943	0.0862
H23_51_A	0.008	7.0 : 1121.0, 7.0 : 612.4	0.006, 0.011	1.306	0.2531
H24_51_A	0.007	8.2 : 1119.8, 4.8 : 614.6	0.007, 0.008	0.01	0.9188
H25_51_A	0.007	10.2 : 1117.8, 2.3 : 617.1	0.009, 0.004	1.655	0.1983
H26_51_A	0.007	9.3 : 1118.7, 3.0 : 616.4	0.008, 0.005	0.653	0.4191
H27_51_A	0.007	7.1 : 1120.9, 5.0 : 614.4	0.006, 0.008	0.183	0.6691
H28_51_A	0.007	10.6 : 1117.4, 1.3 : 618.1	0.009, 0.002	3.177	0.0747
H29_51_A	0.007	4.0 : 1123.9, 7.8 : 611.6	0.004, 0.013	4.885	0.0271
H30_51_A	0.006	8.0 : 1119.9, 3.0 : 616.4	0.007, 0.005	0.315	0.5745
H31_51_A	0.006	6.7 : 1121.2, 3.3 : 616.1	0.006, 0.005	0.034	0.8537
H32_51_A	0.005	4.0 : 1124.0, 5.8 : 613.6	0.004, 0.009	2.383	0.1227

FIG. 22 CONT.

H33_51_A	0.005	5.0 : 1123.0, 3.9 : 615.5	0.004, 0.006	0.259	0.6106
H34_51_A	0.005	4.0 : 1124.0, 4.8 : 614.6	0.004, 0.008	1.419	0.2336
H35_51_A	0.004	3.0 : 1125.0, 4.0 : 615.4	0.003, 0.006	1.446	0.2292
H36_51_A	0.004	2.6 : 1125.4, 4.0 : 615.4	0.002, 0.006	1.82	0.1774
H37_51_A	0.003	5.0 : 1122.9, 1.1 : 618.3	0.004, 0.002	0.774	0.3791
H38_51_A	0.003	3.0 : 1124.9, 2.5 : 616.9	0.003, 0.004	0.242	0.6229
H39_51_A	0.003	3.1 : 1124.9, 2.0 : 617.4	0.003, 0.003	0.034	0.854
H40_51_A	0.003	2.0 : 1125.9, 3.0 : 616.4	0.002, 0.005	1.274	0.259
H41_51_A	0.003	3.0 : 1125.0, 2.0 : 617.4	0.003, 0.003	0.045	0.8312
H42_51_A	0.003	2.0 : 1125.9, 2.7 : 616.7	0.002, 0.004	0.925	0.3362
H43_51_A	0.002	3.4 : 1124.5, 1.0 : 618.4	0.003, 0.002	0.343	0.5578
H44_51_A	0.002	1.0 : 1127.0, 3.1 : 616.3	0.001, 0.005	2.843	0.0918
H45_51_A	0.002	4.0 : 1124.0, 0.0 : 619.4	0.004, 0.000	2.202	0.1379
H46_51_A	0.002	2.2 : 1125.8, 1.0 : 618.4	0.002, 0.002	0.016	0.8993
H47_51_A	0.002	2.0 : 1125.9, 1.0 : 618.4	0.002, 0.002	0.003	0.9578
H48_51_A	0.002	2.0 : 1126.0, 1.0 : 618.4	0.002, 0.002	0.006	0.9389
H49_51_A	0.002	0.0 : 1128.0, 3.0 : 616.4	0.000, 0.005	5.46	0.0195
H50_51_A	0.001	2.4 : 1125.6, 0.2 : 619.2	0.002, 0.000	0.973	0.3239
H51_51_A	0.001	2.4 : 1125.6, 0.0 : 619.4	0.002, 0.000	1.316	0.2513
H52_51_A	0.001	2.1 : 1125.9, 0.2 : 619.2	0.002, 0.000	0.762	0.3826
H53_51_A	0.001	1.0 : 1127.0, 1.1 : 618.3	0.001, 0.002	0.245	0.6206
H54_51_A	0.001	2.0 : 1126.0, 0.1 : 619.3	0.002, 0.000	0.917	0.3381
H55_51_A	0.001	1.0 : 1127.0, 1.0 : 618.4	0.001, 0.002	0.191	0.6618
H56_51_A	0.001	2.0 : 1126.0, 0.0 : 619.4	0.002, 0.000	1.076	0.2997
H57_51_A	0.001	0.0 : 1128.0, 2.0 : 617.4	0.000, 0.003	3.645	0.0562
H58_51_A	0.001	1.0 : 1127.0, 1.0 : 618.4	0.001, 0.002	0.185	0.6669
H59_51_A	0.001	0.0 : 1128.0, 2.0 : 617.4	0.000, 0.003	3.646	0.0562
H60_51_A	0.001	2.0 : 1126.0, 0.0 : 619.4	0.002, 0.000	1.1	0.2944
H61_51_A	0.001	2.0 : 1126.0, 0.0 : 619.4	0.002, 0.000	1.1	0.2944
H62_51_A	0.001	1.0 : 1127.0, 1.0 : 618.4	0.001, 0.002	0.185	0.6669
H63_51_A	0.001	1.9 : 1126.0, 0.0 : 619.4	0.002, 0.000	1.03	0.3102
H64_51_A	0.001	0.0 : 1128.0, 1.9 : 617.5	0.000, 0.003	3.493	0.0616
H65_51_A	0.001	1.9 : 1126.1, 0.0 : 619.4	0.002, 0.000	1.009	0.3151

FIG. 22 CONT.

For 2x2 for only female subjects									
	fem case	fem non case	fem control	fem non control	Pearson Chi2	p-value	OR	lower 95% CI	upper 95% CI
H1_62_A	177	575	57	295	7.740	0.005	1.593	1.146	2.216
H2_62_A	154	598	29	323	25.980	<.0001	2.868	1.886	4.362
H3_62_A	37	715	41	311	16.530	<.0001	0.393	0.247	0.624
H4_62_A	50	702	23	329	0.010	0.920	1.019	0.611	1.698
H5_62_A	25	727	33	319	17.630	<.0001	0.332	0.195	0.568
H6_62_A	20	732	18	334	4.340	0.037	0.507	0.265	0.971
H7_62_A	22	730	13	339	0.460	0.498	0.786	0.391	1.579
H8_62_A	17	735	8	344	0.000	1.000	0.995	0.425	2.327
H9_62_A	19	733	7	345	0.300	0.584	1.278	0.532	3.068
H10_62_A	11	741	4	348	na	na	1.292	0.408	4.085
H11_62_A	3	749	6	346	na	na	0.231	0.057	0.929
H12_62_A	12	740	5	347	0.050	0.823	1.125	0.393	3.219
H13_62_A	5	747	6	346	na	na	0.386	0.117	1.273
H14_62_A	7	745	4	348	na	na	0.817	0.238	2.811
H15_62_A	8	744	3	349	na	na	1.251	0.330	4.744

FIG. 23

For 2x2 for only male subjects									
	male case	Male non case	Male control	male non control	Pearson Chi2	p-value	OR	lower 95% CI	upper 95% CI
H1_62_A	74	370	60	246	1.070	0.301	0.820	0.563	1.195
H2_62_A	76	368	32	274	6.520	0.011	1.768	1.137	2.750
H3_62_A	30	414	30	276	2.290	0.130	0.667	0.393	1.131
H4_62_A	32	412	11	295	4.370	0.037	2.083	1.033	4.200
H5_62_A	23	421	20	286	0.620	0.431	0.781	0.421	1.449
H6_62_A	18	426	11	295	0.100	0.752	1.133	0.528	2.434
H7_62_A	17	427	10	296	0.160	0.689	1.179	0.532	2.610
H8_62_A	10	434	7	299	0.000	1.000	0.984	0.371	2.615
H9_62_A	5	439	4	302	na	na	0.860	0.229	3.229
H10_62_A	10	434	5	301	0.350	0.554	1.387	0.469	4.099
H11_62_A	5	439	15	291	9.950	0.002	0.221	0.079	0.615
H12_62_A	7	437	5	301	na	na	0.964	0.303	3.067
H13_62_A	4	440	7	299	na	na	0.388	0.113	1.338
H14_62_A	8	436	5	301	0.030	0.862	1.105	0.358	3.409
H15_62_A	6	438	3	303	na	na	1.394	0.343	5.575

FIG. 23 CONT.

Haplotype	Haplotype Sequence Male Subjects Only	r35928059	r35753394	r551397	r5000292	r3766404	r16840422	r1061147	r10801554	r1061170	r12124794	r2274700	r3753395	r393995	r403046	r1410996	r12144939	r2284664	r355292076	r1065489	r11562939	r13876 (r112797613)	r12406509 (mrd_3070)	r12066959	r3828032	r12_3902 (r79351096)	r12_3905 (r75816959)	r12_3906 (r74180363)	r56702340	r3748557	r12097550	r1750311	r698859	r5998	r2990510	r10801586	r17514281	r5997	r1615413	r1412631 (mrd_3922)	r6428380 (r115247)					
H1_51_B	ACCGATCAACCATGACCAAGGCGCGGACGCGGGGTTCGGGCTTCGTCC	A	C	C	G	A	T	C	A	C	C	A	T	G	A	C	A	G	C	C	C	C	C	G	G	C	G	G	G	G	T	G	C	G	C	T	C	C								
H2_51_B	ACCGATCAACCATGACCAAGGCGCGGACGCGGGGTTCGGATCTTCATCC	A	C	C	G	A	T	C	A	C	C	A	T	G	A	C	G	C	C	C	C	C	C	G	G	C	G	G	G	G	T	G	C	G	T	A	C	T	C							
H3_51_B	ACTAATCCTTTTCATAAAGTTCGCGGATACGGGGTTCGTGTTTCGTCC	A	C	T	A	T	C	C	T	T	C	A	T	A	G	A	G	T	C	G	C	G	C	A	C	G	G	G	G	G	T	G	C	T	C	G	T	T	C	G	T	C				
H4_51_B	ATCGATCCTTTACGAAGGGGCTTCGAATGCAGAGTTCCTTCATCC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	G	G	C	C	T	C	G	A	T	G	C	A	A	A	G	T	A	G	A	T	A	T	C	T	C	A	T	C			
H5_51_B	ACGGGCTCTTTATTAAGATTCGCGGGATGCAGGCTTCGATCATCTTACTT	A	C	C	G	G	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	G	T	C	T	G	A	T	C	A	T	T	C	A	T			
H6_51_B	ACTAATCCTTTTCATAAAGTTCGCGGATACGAAGTAGATTATCTTCATCC	A	C	T	A	T	C	C	T	T	C	A	T	A	A	G	A	G	T	C	G	C	G	A	T	A	C	A	A	G	T	A	G	A	T	A	T	C	T	C	A	T	C			
H7_51_B	GTCCGATCCTTTATGACAGGCGCGGACGCGGGGTTCGGATCTTCATCC	G	T	C	G	A	T	C	C	T	T	A	T	G	A	C	A	G	C	C	G	C	G	A	C	G	C	G	G	G	T	G	C	G	T	A	T	C	T	C	A	T	C			
H8_51_B	ACCGATCAACCATGACCAAGGCGCGGACGCGAGGTAGATTATCTTCATCC	A	C	C	G	A	T	C	A	C	A	C	A	T	G	A	C	A	G	C	C	G	C	G	A	C	G	C	A	G	T	A	G	A	T	A	T	A	T	C	T	C	A	T	C	
H9_51_B	ATCGATCAACCATGACCAAGGCGCGGACGCGGGGTTCGGGCTTCGTCC	A	T	C	G	A	T	C	A	C	A	C	A	T	G	A	C	A	G	C	C	G	C	G	A	C	G	C	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C	C		
H10_51_B	ATCGATCCTTTATTAAGATTCGCGGAATGCAGAGTAGATTATCTTCATCC	A	T	C	G	A	T	C	C	T	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	A	A	G	T	A	G	A	T	A	T	C	T	C	A	T	C	C	
H11_51_B	ACCGATCAACCATGACCAAGGCTTCGCTGACGCGGGGTTCGGGCTTCGTCC	A	C	C	G	A	T	C	A	C	A	C	A	T	G	A	C	A	G	C	T	G	C	T	G	A	C	G	C	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C	C	
H12_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTTCAGGGTTCGGGCTTCGTCC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	G	G	G	C	C	T	C	G	A	G	T	G	C	A	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C	C	
H13_51_B	ACCGATCAACCATGACCAAGGCGCGGATTCGGGTTTCGGGCTTCGTCC	A	C	C	G	A	T	C	A	C	A	C	A	T	G	A	C	A	G	C	C	G	C	G	A	T	G	C	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C	C		
H14_51_B	ACGGGCTCTTTATTAAGATTCGCGGGATGCAGAGTTCGATCATCC	A	C	C	G	G	C	T	C	T	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	A	A	G	T	A	G	A	T	A	T	C	T	C	A	T	C	C	
H15_51_B	ATCGGCTCTTTATTAAGATTCGCGGGATGCAGGCTCGATCATCTTACTT	A	T	C	G	G	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	G	T	C	T	G	A	T	C	A	T	T	T	A	C	T	T	
H16_51_B	ATCGATCCTTTACGAAGGGGCTTCGGATTCGGGGTTTCGGGCTTCGTCC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	G	G	G	C	C	T	C	G	G	A	T	G	C	G	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C	C	
H17_51_B	ACCGATCAACCATGACCAAGGCGCGGATACGGGGTTCGTGTTTCGTCC	A	C	C	G	A	T	C	A	C	A	C	A	T	G	A	C	A	G	C	C	G	C	G	A	T	A	C	G	G	T	T	G	C	T	C	G	T	T	T	C	G	T	C	C	
H18_51_B	ACGGGCTCTTTATTAAGATTCGCGGGATGCAGGGTTCGGGCTTCGTCC	A	C	C	G	G	C	T	C	T	T	A	T	A	A	G	A	T	C	C	G	C	G	G	A	T	G	C	A	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C	C	
H19_51_B	ACCGATCAACCATGACCAAGGCGCGGACGCGGGGTTCGGGCTTCGTCC	A	C	C	G	A	T	C	A	C	A	C	A	T	G	A	C	A	G	C	C	G	C	G	A	C	G	C	G	G	T	T	A	C	G	C	G	C	T	T	A	C	G	T	C	C

FIG. 24A

Haplotype	Freq.	Case, Control Ratio Counts	Case,Control Freqs	Chi Square	P Value
H1_51_B	0.219	475.3 : 1380.8, 134.1 : 687.4	0.256, 0.163	27.895	1.28E-07
H2_51_B	0.144	318.7 : 1537.3, 81.8 : 739.7	0.172, 0.100	23.278	1.40E-06
H3_51_B	0.087	138.0 : 1718.0, 103.6 : 717.9	0.074, 0.126	18.554	1.65E-05
H4_51_B	0.061	113.1 : 1742.9, 55.4 : 766.1	0.061, 0.067	0.411	0.5216
H5_51_B	0.051	84.0 : 1772.1, 58.9 : 762.7	0.045, 0.072	7.861	0.0051
H6_51_B	0.036	59.8 : 1796.2, 39.6 : 782.0	0.032, 0.048	4.047	0.0443
H7_51_B	0.03	57.9 : 1798.1, 26.6 : 795.0	0.031, 0.032	0.024	0.8758
H8_51_B	0.021	38.5 : 1817.5, 19.0 : 802.5	0.021, 0.023	0.156	0.6931
H9_51_B	0.02	39.5 : 1816.6, 16.2 : 805.3	0.021, 0.020	0.065	0.798
H10_51_B	0.018	25.0 : 1831.1, 24.8 : 796.8	0.013, 0.030	8.713	0.0032
H11_51_B	0.016	35.4 : 1820.6, 8.9 : 812.6	0.019, 0.011	2.378	0.123
H12_51_B	0.015	28.4 : 1827.6, 14.0 : 807.6	0.015, 0.017	0.106	0.7442
H13_51_B	0.014	30.0 : 1826.0, 10.2 : 811.3	0.016, 0.012	0.542	0.4614
H14_51_B	0.014	17.6 : 1838.4, 21.5 : 800.0	0.009, 0.026	11.063	9.00E-04
H15_51_B	0.013	23.0 : 1833.0, 14.1 : 807.4	0.012, 0.017	0.959	0.3275
H16_51_B	0.013	23.0 : 1833.1, 12.7 : 808.8	0.012, 0.015	0.415	0.5195
H17_51_B	0.012	28.0 : 1828.0, 6.4 : 815.1	0.015, 0.008	2.397	0.1216
H18_51_B	0.012	16.8 : 1839.2, 15.4 : 806.1	0.009, 0.019	4.466	0.0346
H19_51_B	0.01	23.1 : 1832.9, 5.0 : 816.5	0.012, 0.006	2.251	0.1336

FIG. 24A CONT.

Haplotype	Haplotype Sequence Female Subjects Only	r3359228059	r33753394	r5551397	r5800292	r3766404	r168440422	r1061147	r13329421	r10801554	r1061170	r12124794	r22274700	r33753395	r33753396	r403846	r1410996	r12144939	r2284664	r35292876	r1065489	r11582939	mrd_3876 (r112797613)	mrd_3877	r7546940	r12046509 (mrd_3878)	r1409153	r12066959	mrd_3902 (r79351096)	r3828032	mrd_3905 (r75816959)	mrd_3906 (r74188363)	r6702340	r3748557	r12097550	r1750311	r10922153	r698859	r5998	r2990510	r5991	r17514281	r5997	r1615413	r6003	r1412631 (mrd_3922)	r6428380 (r115247)			
41_51_B	AACGATCAACCATGACAAAGGCGCGGACGCGGGGTTGCGGGCTTGTC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	G	G	C	C	C	C	C	C	G	G	A	C	G	G	G	G	G	G	G	T	G	C	G	C	G	C	T	C	C	C				
42_51_B	AACGATCAACCATGACAAAGGCGCGGACGCGGGGTTGCGTATCTTCATCC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	G	G	G	C	C	C	C	C	C	G	G	A	C	G	G	G	G	G	G	T	G	C	G	C	G	C	T	C	C					
43_51_B	ACTAATCCTTTTCATAAAGTTCGCGGATACGGGGTGCTGGTTTCGTC	A	C	T	A	T	C	C	T	T	T	C	A	T	A	A	A	G	A	G	T	C	G	C	C	C	G	G	A	T	A	C	G	G	G	G	T	G	C	T	T	C	G	T	C	C				
44_51_B	ATCGATCCTTTACGAAGGGGCTTCGAATGCGAAAGTAGATATCTTCATCC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	C	G	A	T	G	C	A	A	A	G	T	A	G	A	T	A	G	A	T	A	T	C	T	C	C			
45_51_B	ACCGGCTCTTTATATAGATTCGCGCGGATGCAGGCTGTGATCATCTTTACTT	A	C	C	G	G	C	T	C	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	G	T	C	A	G	T	C	T	G	A	T	C	T	A	C	T				
46_51_B	ACTAATCCTTTTCATAAAGTTCGCGGATACGAAGTAGATTATCTTCATCC	A	C	T	A	T	C	C	T	T	T	C	A	T	A	A	A	G	A	G	T	C	G	C	C	G	A	T	A	C	G	A	A	G	T	A	G	A	T	A	T	A	T	C	T	C	C			
47_51_B	GTGATCCTTTATGACAAGGCGCGGACGCGGGGTTGGTATCTTCATCC	G	T	C	G	A	T	C	C	T	T	A	T	G	A	C	A	G	C	C	G	C	C	C	C	G	A	C	G	C	G	G	G	T	G	C	G	T	A	T	A	T	C	T	C	C				
48_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTGCAGGGTTGCGGGCTTGTC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	C	G	A	T	G	A	G	T	G	C	A	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C		
49_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTGCAGGGTTGCGGGCTTGTC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	C	G	A	T	G	A	G	T	G	C	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C			
410_51_B	AACGATCAACCATGACAAAGGCGCGGACGCGGAAGTAGATTATCTTCATCC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	C	C	C	C	C	G	A	C	G	C	G	A	A	G	T	A	G	A	T	A	T	A	T	C	T	C	C			
411_51_B	ATCGATCAACCATGACAAAGGCGCGGACGCGGGGTTGCGGGGCTTGTC	A	T	C	G	A	T	C	A	C	A	C	A	T	G	A	C	A	G	C	C	G	C	C	G	A	C	G	C	G	G	T	T	G	C	G	T	T	G	C	G	C	T	C	G	T	C			
412_51_B	ATCGATCCTTTATAGATTCGCGGATGCAAGTAGATTATCTTCATCC	A	T	C	G	A	T	C	C	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	A	A	G	T	A	G	A	T	A	T	C	T	C	T	C	A	T	C		
413_51_B	AACGATCAACCATGACAAAGGCTGCTGACGCGGGGTTGCGGGGCTTGTC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	T	G	C	C	T	G	A	C	G	C	G	G	T	T	G	C	G	C	G	C	T	T	G	C	T	C	C			
414_51_B	ACCGGCTCTTTATATAGATTCGCGCGGATGCAGGCTGTGATCATCTTCATCC	A	C	C	G	G	C	T	C	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	A	A	G	T	A	G	A	T	A	T	A	T	C	T	C	A	T	C		
415_51_B	ACCGGCTCTTTATATAGATTCGCGCGGATGCAGGCTGTGCGGGCTTGTC	A	C	C	G	G	C	T	C	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	G	G	T	T	G	C	G	C	G	C	T	T	G	C	T	C	C			
416_51_B	AACGATCAACCATGACAAAGGCGCGGACGCGGGGTTGCTGGTTTCGTC	A	C	C	G	A	T	C	A	C	A	T	G	A	C	A	A	G	C	C	G	C	C	C	C	G	A	T	A	C	G	G	G	T	T	G	C	T	T	G	C	T	T	C	G	T	C	C		
417_51_B	ATCGGCTCTTTATATAGATTCGCGCGGATGCAGGCTGTGATCATCTTTACTT	A	T	C	G	G	C	T	C	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	G	A	T	G	C	A	G	T	C	A	G	T	T	G	A	T	C	T	T	A	C	T	T			
418_51_B	ATCGATCCTTTACGAAGGGGCTTCGATAGCGGGTTGCTGGTTTCGTC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	C	G	A	T	A	C	G	G	G	T	T	G	C	T	T	G	C	T	T	T	C	G	T	C	C			
419_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTGTGGGGTTGCGGGGCTTGTC	A	T	C	G	A	T	C	C	T	T	A	C	G	A	A	G	G	G	C	C	T	C	G	A	T	G	G	A	T	G	T	G	G	G	T	T	G	C	G	C	G	C	T	C	G	T	C		
420_51_B	AACGATCAACCATGACAAAGGCGCGGATGCAAGTAGATTATCTTCATCC	A	C	C	G	A	T	C	A	C	A	C	A	T	G	A	C	A	G	C	C	G	C	C	G	A	T	G	C	A	A	A	G	T	A	G	A	T	A	T	A	T	A	T	C	T	C	A	T	C

FIG. 24B

Haplotype	Freq.	Case, Control Ratio Counts	Case,Control Freqs	Chi Square	P Value
H1_51_B	0.212	773.5 : 2547.2, 177.9 : 802.6	0.233, 0.181	11.661	6.00E-04
H2_51_B	0.155	589.3 : 2731.4, 106.2 : 874.3	0.177, 0.108	26.708	2.37E-07
H3_51_B	0.081	223.1 : 3097.6, 142.5 : 838.0	0.067, 0.145	59.498	1.22E-14
H4_51_B	0.075	251.4 : 3069.3, 84.6 : 895.9	0.076, 0.086	1.183	0.2768
H5_51_B	0.05	145.9 : 3174.8, 80.1 : 900.4	0.044, 0.082	21.729	3.14E-06
H6_51_B	0.04	133.7 : 3186.9, 45.3 : 935.3	0.040, 0.046	0.658	0.4172
H7_51_B	0.026	90.5 : 3230.2, 27.2 : 953.4	0.027, 0.028	0.006	0.9379
H8_51_B	0.021	75.1 : 3245.6, 17.9 : 962.6	0.023, 0.018	0.676	0.4109
H9_51_B	0.019	70.3 : 3250.3, 15.2 : 965.3	0.021, 0.016	1.251	0.2634
H10_51_B	0.018	66.3 : 3254.4, 15.5 : 965.0	0.020, 0.016	0.696	0.4041
H11_51_B	0.016	61.8 : 3258.9, 10.0 : 970.5	0.019, 0.010	3.267	0.0707
H12_51_B	0.016	42.5 : 3278.2, 28.5 : 952.1	0.013, 0.029	12.301	5.00E-04
H13_51_B	0.014	53.8 : 3266.8, 7.0 : 973.5	0.016, 0.007	4.47	0.0345
H14_51_B	0.012	31.5 : 3289.1, 24.7 : 955.8	0.009, 0.025	14.405	1.00E-04
H15_51_B	0.012	36.4 : 3284.3, 16.5 : 964.0	0.011, 0.017	2.17	0.1407
H16_51_B	0.011	44.4 : 3276.2, 5.3 : 975.2	0.013, 0.005	4.243	0.0394
H17_51_B	0.011	30.8 : 3289.8, 17.7 : 962.8	0.009, 0.018	5.243	0.022
H18_51_B	0.011	39.6 : 3281.1, 8.9 : 971.6	0.012, 0.009	0.54	0.4623
H19_51_B	0.011	41.9 : 3278.8, 6.3 : 974.3	0.013, 0.006	2.661	0.1029
H20_51_B	0.011	34.4 : 3286.2, 13.2 : 967.3	0.010, 0.013	0.669	0.4133

FIG. 24B CONT.

Haplotype	62_A	51_A	51_B
H1	Neutral	Neutral	Risk
H2	Risk	Risk	Risk
H3	Protective	Protective	Protective
H4	Neutral	Neutral	Neutral
H5	Protective	Protective	Protective
H6	Neutral	Neutral	Neutral
H7	Neutral	Neutral	Neutral
H8	Neutral	Neutral	Neutral
H9	Neutral	Neutral	Neutral
H10	Neutral	Protective	Neutral
H11	Protective	Neutral	Neutral
H12	Neutral	Neutral	Protective
H13	Marginally Prot.	Neutral	Neutral
H14	Neutral	Neutral	Protective
H15	Neutral	Neutral	Neutral
H16		Neutral	Neutral
H17		Neutral	Neutral
H18		Neutral	Neutral
H19		Neutral	Neutral
H20			Neutral
H21			Neutral
H22			Neutral

FIG. 25

Haplotype	62_A Female Only	62_A Male Only	51_B Female Only	51_B Male Only
H1	Risk	Neutral	Risk	Risk
H2	Risk	Risk	Risk	Risk
H3	Protective	Neutral	Protective	Protective
H4	Neutral	Neutral	Neutral	Neutral
H5	Protective	Neutral	Protective	Neutral
H6	Neutral	Neutral	Neutral	Neutral
H7	Neutral	Neutral	Neutral	Neutral
H8	Neutral	Neutral	Neutral	Neutral
H9	Neutral	Neutral	Neutral	Neutral
H10	Neutral	Neutral	Neutral	Neutral
H11	Protective	Protective	Neutral	Neutral
H12	Neutral	Neutral	Protective	Neutral
H13	Neutral	Neutral	Neutral	Neutral
H14	Neutral	Neutral	Protective	Protective
H15	Neutral	Neutral	Neutral	Neutral
H16			Neutral	Neutral
H17			Neutral	Neutral
H18			Neutral	Neutral
H19			Neutral	Neutral
H20			Neutral	

FIG. 26

SNPs used in haplotype reconstruction				
SNP	Position ^a	Major Allele	Minor Allele	MAF ^b (%)
mrd_3876	195123565	C	G	1.2
mrd_3877	195123773	C	A	1.0
mrd_3902	195185364	G	A	1.8
mrd_3905	195211345	G	A	19.5
mrd_3906	195211483	C	T	19.8

^aPositions are relative to the start of chromosome 1 in NCBI build 36 of the reference human genome

^bMinor allele frequency

FIG. 27

TaqMan qPCR Copy Number Variation Assay

Genotype	Count
NN	387
ND	133
DD	16
N? ^a	537

^aPartially known genotypes are based on ambiguous qPCR or the presence of known SNPs in the CFHR-3 / CFHR-1 genes.

FIG. 28

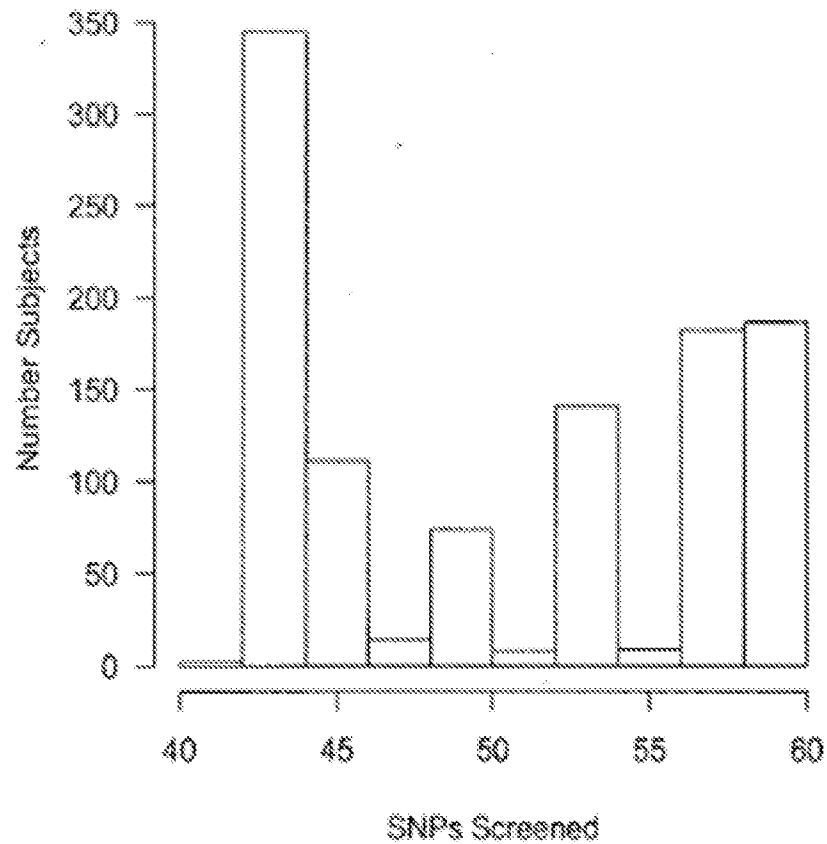


FIG. 29

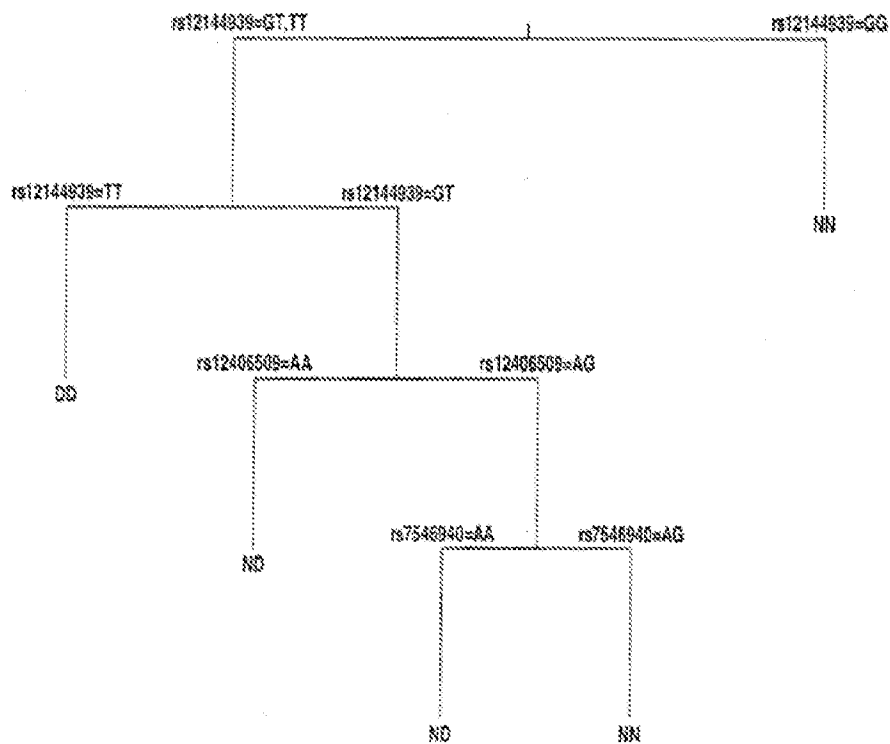


FIG. 30

qPCR Deletion Assay

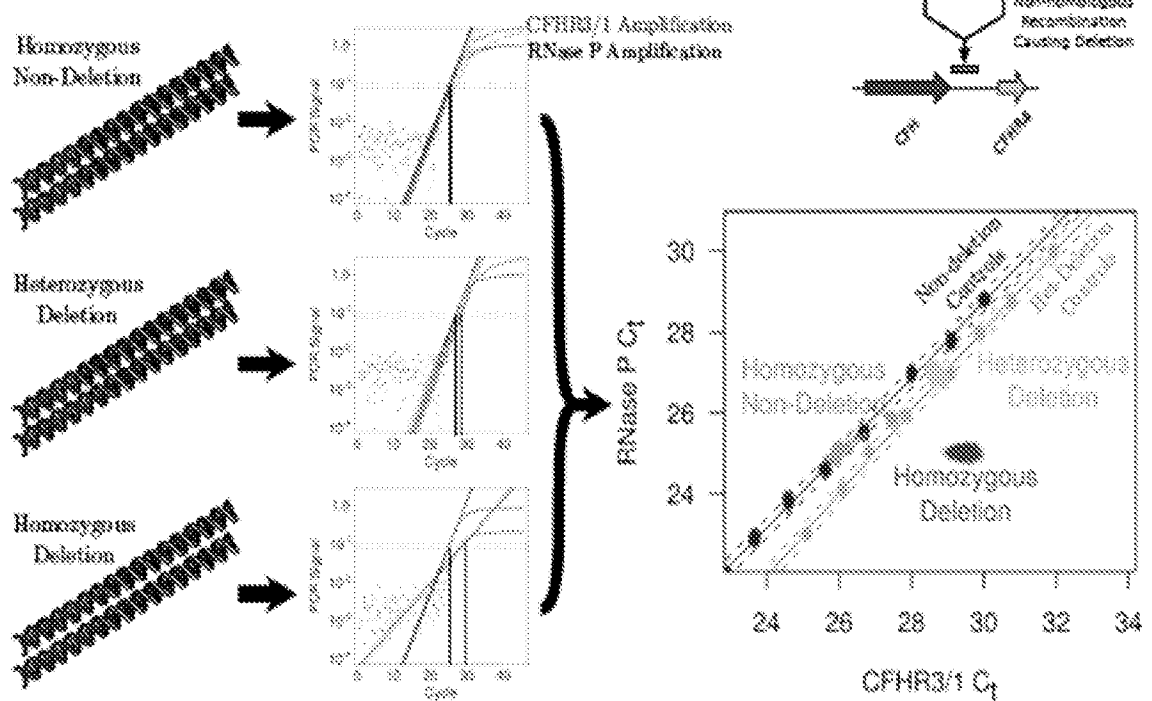


FIG. 31

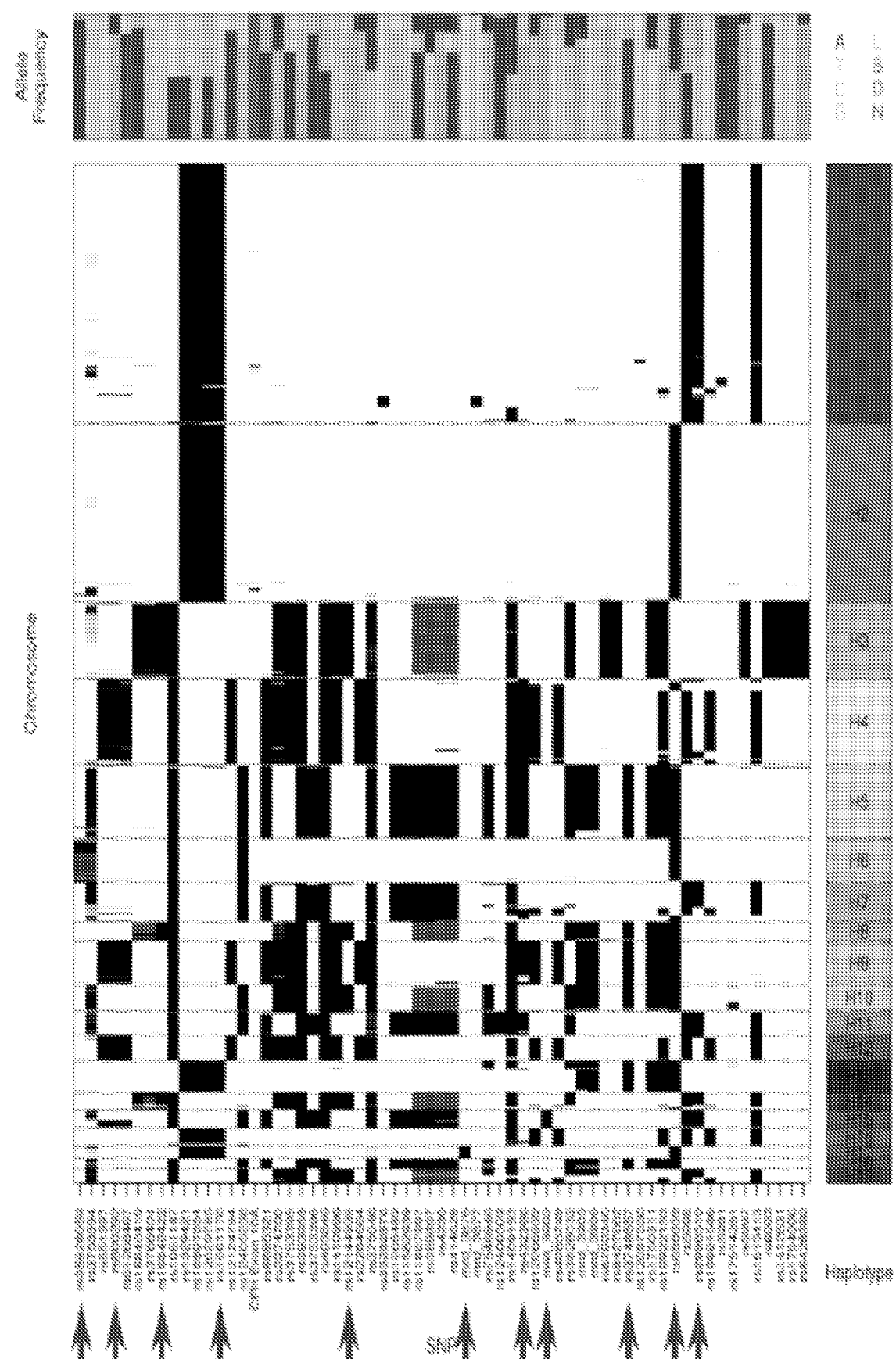


FIG. 32

[illegible]

boxes mark haplotypes with *CFHR3/1* gene deletions and are tagged by the rs12144930 SNP.

FIG. 33

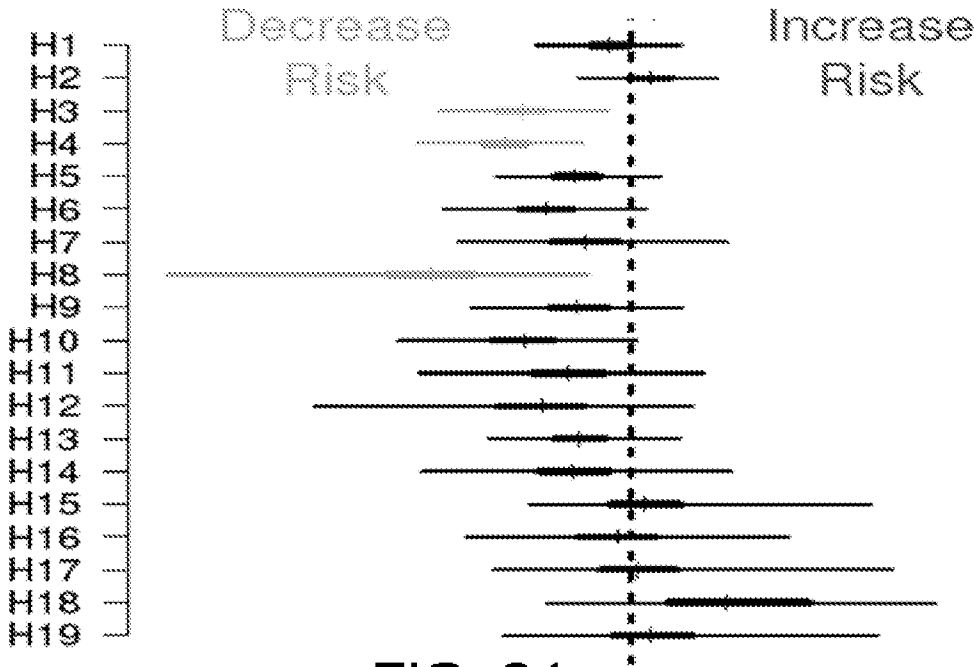


FIG. 34

Diplotype	Total	Cases with	Cases w/o	Controls with	Controls w/o	p-value	Chi Square (Pearson)	Risk Ratio	Odds Ratio	95% CI lower	95% CI upper
H1_51_B:H1_51_B	179	153	2502	26	1047	<.0001	18.65	1.0354	2.4625	1.6148	3.7553
H1_51_B:H2_51_B	272	226	2429	46	1027	<.0001	20.17	1.0462	2.0773	1.5004	2.8759
H1_51_B:H3_51_B	120	71	2584	49	1024	0.003046	8.78	0.9806	0.5742	0.3362	0.8323
H1_51_B:H4_51_B	94	69	2586	25	1048	0.63904	0.22	1.0028	1.1185	0.7039	1.7774
H1_51_B:H5_51_B	76	44	2611	32	1041	0.009534	6.72	0.9865	0.5482	0.3457	0.8893
H1_51_B:H6_51_B	58	40	2615	18	1055	0.698535	0.15	0.9983	0.8965	0.5117	1.5709
H1_51_B:H7_51_B	38	31	2624	7	1066	0.156265	2.01	1.0052	1.7991	0.7898	4.0983
H1_51_B:H8_51_B	41	36	2619	5	1068	0.018375	5.56	1.009	2.9361	1.1491	7.5023
H1_51_B:H9_51_B	35	27	2628	8	1065	0.434788	0.61	1.0027	1.3677	0.6194	3.0202
H1_51_B:H10_51_B	31	27	2628	4	1069	0.049746	3.85	1.0065	2.7457	0.9584	7.8659
H1_51_B:H11_51_B	28	22	2633	6	1067	0.389661	0.74	1.0027	1.4859	0.6008	3.6749
H1_51_B:H12_51_B	21	16	2639	5	1068	0.617075	0.25	1.0014	1.295	0.4732	3.544
H1_51_B:H13_51_B	22	21	2634	1	1072	0.011804	6.34	1.007	8.5467	1.1482	63.6198
H1_51_B:H14_51_B	27	13	2642	14	1059	0.007882	7.06	0.9918	0.3722	0.1744	0.7945
H1_51_B:H15_51_B	19	14	2641	5	1068	0.806496	0.06	1.0006	1.1323	0.4068	3.1514
H1_51_B:H16_51_B	18	12	2643	6	1067	0.671373	0.18	0.9989	0.8074	0.3023	2.1569
H1_51_B:H17_51_B	18	16	2639	2	1071	0.096648	2.76	1.0042	3.2467	0.7452	14.1448
H1_51_B:H18_51_B	11	8	2647	3	1070			1.0002	1.0779	0.2854	4.071
H1_51_B:H19_51_B	16	11	2644	5	1068			0.9995	0.8887	0.308	2.5638
H1_51_B:H20_51_B	14	11	2644	3	1070			1.0014	1.4839	0.4131	5.3294
H1_51_B:H21_51_B	14	10	2645	4	1069			1	1.0104	0.3162	3.2287
H1_51_B:H22_51_B	12	8	2647	4	1069			0.9993	0.8077	0.2427	2.688
H1_51_B:H23_51_B	13	11	2644	2	1071			1.0023	2.2279	0.493	10.0682
H1_51_B:H24_51_B	10	8	2647	2	1071			1.0012	1.6184	0.3431	7.6339
H1_51_B: any	1394	1068	1587	326	747	<.0001	31.63	1.1647	1.542	1.3253	1.7942

FIG. 35

H2_51_B:H2_51_B	81	74	2581	7	1066	<.0001	15.38	1.022	4.3662	2.0048	9.5089
H2_51_B:H3_51_B	73	54	2601	19	1054	0.596701	0.28	1.0027	1.1517	0.6795	1.9521
H2_51_B:H4_51_B	75	56	2599	19	1054	0.507122	0.44	1.0035	1.1953	0.7069	2.0211
H2_51_B:H5_51_B	56	41	2614	15	1058	0.740144	0.11	1.0015	1.1063	0.6097	2.0072
H2_51_B:H6_51_B	41	34	2621	7	1066	0.096046	2.77	1.0064	1.9755	0.873	4.4701
H2_51_B:H7_51_B	37	30	2625	7	1066	0.183382	1.77	1.0048	1.7404	0.7621	3.9745
H2_51_B:H8_51_B	20	15	2640	5	1068	0.708281	0.14	1.001	1.2136	0.44	3.3476
H2_51_B:H9_51_B	18	15	2640	3	1070	0.254213	1.3	1.0029	2.0265	0.5855	7.0144
H2_51_B:H10_51_B	24	23	2632	1	1072	0.007538	7.14	1.0078	9.3678	1.2635	69.4558
H2_51_B:H11_51_B	13	11	2644	2	1071			1.0023	2.2279	0.493	10.0682
H2_51_B:H12_51_B	16	7	2648	9	1064			0.9942	0.3125	0.1161	0.8413
H2_51_B:H13_51_B	16	12	2643	4	1069			1.0008	1.2134	0.3905	3.7707
H2_51_B:H14_51_B	15	10	2645	5	1068			0.9991	0.8076	0.2754	2.3682
H2_51_B:H15_51_B	11	8	2647	3	1070			1.0002	1.0779	0.2854	4.071
H2_51_B:H16_51_B	18	9	2646	9	1064	0.046318	3.97	0.995	0.4021	0.1592	1.0158
H2_51_B:H17_51_B	13	13	2642	0	1073			1.0049	Infinity	NaN	Infinity
H2_51_B:H18_51_B	7	5	2650	2	1071			1	1.0104	0.1957	5.2159
H2_51_B:H19_51_B	9	9	2646	0	1073			1.0034	Infinity	NaN	Infinity
H2_51_B:H20_51_B	14	10	2645	4	1069			1	1.0104	0.3162	3.2287
H2_51_B:H21_51_B	16	10	2645	6	1067			0.9982	0.6723	0.2437	1.8545
H2_51_B:H22_51_B	14	11	2644	3	1070			1.0014	1.4839	0.4131	5.3294
H2_51_B:H23_51_B	12	11	2644	1	1072			1.0032	4.4599	0.5751	34.5882
H2_51_B:H24_51_B	14	13	2642	1	1072			1.004	5.2748	0.6892	40.3732
H2_51_Bary	1039	824	1831	215	858	<.0001	45.98	1.1595	1.7959	1.5142	2.13

FIG. 35 CONT.

H3_51_B:H3_51_B	37	18	2637	19	1054	0.002304	9.29	0.989	0.3787	0.1979	0.7243
H3_51_B:H4_51_B	48	23	2632	25	1048	0.00332	12.88	0.9852	0.3663	0.207	0.6483
H3_51_B:H5_51_B	30	16	2639	14	1059	0.022814	4.72	0.9929	0.4586	0.223	0.943
H3_51_B:H6_51_B	19	8	2647	11	1062	0.004943	7.9	0.9927	0.2918	0.117	0.7274
H3_51_B:H7_51_B	17	10	2645	7	1066			0.9972	0.5757	0.2186	1.5165
H3_51_B:H8_51_B	8	3	2652	5	1068			0.9965	0.2416	0.0576	1.0129
H3_51_B:H9_51_B	9	5	2650	4	1069			0.9982	0.5042	0.1351	1.8814
H3_51_B:H10_51_B	12	8	2647	4	1069			0.9993	0.8077	0.2427	2.688
H3_51_B:H11_51_B	12	7	2648	5	1068			0.998	0.5647	0.1788	1.783
H3_51_B:H12_51_B	12	4	2651	8	1065			0.994	0.2009	0.0604	0.6685
H3_51_B:H13_51_B	9	5	2650	4	1069			0.9982	0.5042	0.1351	1.8814
H3_51_B:H14_51_B	11	2	2653	9	1064			0.9924	0.0891	0.0192	0.4132
H3_51_B:H15_51_B	6	2	2653	4	1069			0.997	0.2015	0.0368	1.1016
H3_51_B:ary	581	323	2332	258	815	<.0001	81.96	0.8648	0.4375	0.3647	0.5249
H4_51_B:H4_51_B	23	18	2637	5	1068	0.45426	0.56	1.0021	1.458	0.5399	3.9371
H4_51_B:H5_51_B	27	15	2640	12	1061	0.071423	3.25	0.9944	0.5024	0.2344	1.0768
H4_51_B:H6_51_B	15	14	2641	1	1072			1.0044	5.6827	0.7463	43.27
H4_51_B:H7_51_B	15	11	2644	4	1069			1.0004	1.1119	0.3533	3.4995
H4_51_B:H8_51_B	10	7	2648	3	1070			0.9998	0.9428	0.2434	3.653
H4_51_B:H9_51_B	10	5	2650	5	1068			0.9972	0.403	0.1164	1.395
H4_51_B:H10_51_B	12	8	2647	4	1069			0.9993	0.8077	0.2427	2.688
H4_51_B:H11_51_B	10	7	2648	3	1070			0.9998	0.9428	0.2434	3.653
H4_51_B:ary	506	351	2304	155	918	0.322199	0.98	0.9859	0.9023	0.7358	1.1064

FIG. 35 CONT.

H5_51_B:H5_51_B	8	3	2652	5	1068			0.9965	0.2416	0.0576	1.0129
H5_51_B:H6_51_B	22	13	2642	9	1064	0.207326	1.59	0.9965	0.5817	0.2479	1.3649
H5_51_B:H7_51_B	13	9	2646	4	1069			0.9997	0.909	0.2793	2.9581
H5_51_B:H8_51_B	12	6	2649	6	1067			0.9967	0.4028	0.1296	1.2517
H5_51_B:H9_51_B	5	3	2652	2	1071			0.9993	0.6058	0.1011	3.6305
H5_51_B:H10_51_B	0	0	2655	0	1073			1	NaN	NaN	NaN
H5_51_B:H11_51_B	9	7	2648	2	1071			1.0008	1.4156	0.2936	6.8254
H5_51_B:H12_51_B	9	4	2651	5	1068			0.9968	0.3223	0.0864	1.2025
H5_51_Bany	358	209	2446	149	924	<.0001	31.84	0.9347	0.5299	0.4238	0.6625
H6_51_B:H6_51_B	5	1	2654	4	1069			0.9966	0.1007	0.0112	0.902
H6_51_B:H7_51_B	6	5	2650	1	1072			1.001	2.0226	0.236	17.3336
H6_51_B:H8_51_B	5	4	2651	1	1072			1.0006	1.6175	0.1806	14.4888
H6_51_Bany	277	180	2475	97	976	0.017159	5.68	0.9758	0.7318	0.5655	0.9469
H7_51_B:H7_51_B	2	2	2653	0	1073			1.0008	Infinity	NaN	Infinity
H7_51_B:H8_51_B	4	3	2652	1	1072			1.0002	1.2127	0.126	11.6715
H8_51_B:H8_51_B	2										
H8_51_B:H9_51_B	2										
H10_51_B:H10_51_B	1										
H11_51_B:H11_51_B	3										

FIG. 35 CONT.

Diplotype	Total	Cases with	Cases w/o	Controls with	Controls w/o	p-value	Chi Square (Pearson)	Risk Ratio	Odds Ratio	95% CI lower	95% CI upper
H1_51_B:H1_51_B	74	61	891	13	449	0.004427	8.1	1.0384	2.3646	1.2856	4.3492
H1_51_B:H2_51_B	89	76	876	13	449	0.000174	14.09	1.0562	2.9965	1.6459	5.4554
H1_51_B:H3_51_B	52	31	921	21	441	0.22693	1.46	0.9867	0.7068	0.4016	1.2442
H1_51_B:H4_51_B	33	20	932	13	449	0.406164	0.69	0.9927	0.7412	0.3654	1.5035
H1_51_B:H5_51_B	36	22	930	14	448	0.420113	0.65	0.9926	0.757	0.3837	1.4935
H1_51_B:H6_51_B	20	16	936	4	458	0.223775	1.48	1.0083	1.9573	0.6506	5.8879
H1_51_B:H7_51_B	10	7	945	3	459			1.0009	1.1333	0.2917	4.403
H1_51_B:H8_51_B	20	16	936	4	458	0.223775	1.48	1.0083	1.9573	0.6506	5.8879
H1_51_B:H9_51_B	10	7	945	3	459						
H1_51_B:H10_51_B	18	14	938	4	458						
H1_51_B:H11_51_B	6	4	948	2	460						
H1_51_B:H12_51_B	9	6	946	3	459						
H1_51_B:H13_51_B	9	8	944	1	461						
H1_51_B:H14_51_B	13	6	946	7	455						
H1_51_B:H15_51_B	6	6	946	0	462						
H2_51_B:H2_51_B	28	27	925	1	461	0.000911	11	1.027	13.4562	1.8228	99.3385
H2_51_B:H3_51_B	23	15	937	8	454	0.823063	0.05	0.9984	0.9085	0.3824	2.1585
H2_51_B:H4_51_B	25	18	934	7	455	0.617075	0.25	1.0038	1.2527	0.5195	3.0207
H2_51_B:H5_51_B	29	20	932	9	453	0.841481	0.04	1.0016	1.0801	0.4879	2.3911
H2_51_B:H6_51_B	15	11	941	4	458			1.0029	1.3385	0.4239	4.2264
H2_51_B:H7_51_B	19	15	937	4	458						
H2_51_B:H8_51_B	8	5	947	3	459						
H2_51_B:H9_51_B	8	5	947	3	459						
H2_51_B:H10_51_B	10	9	943	1	461						
H2_51_B:H11_51_B	3	3	949	0	462						
H2_51_B:H12_51_B	6	0	952	6	456						
H2_51_B:H13_51_B	6	6	946	0	462						
H2_51_B:H14_51_B	5	3	949	2	460						
H2_51_B:H15_51_B	2	1	951	1	461						

FIG. 36

H3_51_B:H3_51_B	15	7	945	8	454				0.99	0.4204	0.1515	1.1664
H3_51_B:H4_51_B	20	10	942	10	452	0.096046	2.77		0.9887	0.4798	0.1983	1.1611
H3_51_B:H5_51_B	8	5	947	3	459				0.9988	0.8078	0.1922	3.395
H3_51_B:H6_51_B	8	2	950	6	456				0.9891	0.16	0.0322	0.7958
H3_51_B:H7_51_B	7	5	947	2	460							
H3_51_B:H8_51_B	2	1	951	1	461							
H3_51_B:H9_51_B	1	1	951	0	462							
H3_51_B:H10_51_B	6	3	949	3	459							
H4_51_B:H4_51_B	5	4	948	1	461				1.002	1.9451	0.2168	17.453
H4_51_B:H5_51_B	9	6	946	3	459							
H4_51_B:H6_51_B	2	2	950	0	462							
H4_51_B:H7_51_B	6	5	947	1	461							
H4_51_B:H8_51_B	4	2	950	2	460							
H4_51_B:H9_51_B	5	2	950	3	459							
H4_51_B:H10_51_B	5	3	949	2	460							
H5_51_B:H5_51_B	2	1	951	1	461				0.9989	0.4848	0.0303	7.7677
H5_51_B:H6_51_B	9	5	947	4	458							
H5_51_B:H7_51_B	5	3	949	2	460							
H5_51_B:H8_51_B	5	2	950	3	459							
H5_51_B:H9_51_B	1	0	952	1	461							
H5_51_B:H10_51_B	0	0	952	0	462							
H6_51_B:H6_51_B	2	0	952	2	460							
H6_51_B:H7_51_B	3	2	950	1	461							

FIG. 36 CONT.

Diplotype	Total	Cases with	Cases w/o	Controls with	Controls w/o	p-value	Chi Square (Pearson)	Risk Ratio	Odds Ratio	95% CI lower	95% CI upper
H1_51_B:H1_51_B	105	92	1611	13	598	0.000849	11.13	1.0346	2.6269	1.4587	4.7308
H1_51_B:H2_51_B	183	150	1553	33	578	0.007413	7.17	1.0374	1.6917	1.1468	2.4956
H1_51_B:H3_51_B	68	40	1663	28	583	0.005026	7.87	0.9771	0.5008	0.3062	0.8192
H1_51_B:H4_51_B	61	49	1654	12	599	0.22693	1.46	1.0094	1.4788	0.7812	2.7994
H1_51_B:H5_51_B	40	22	1681	18	593	0.00713	7.24	0.9832	0.4312	0.2296	0.8095
H1_51_B:H6_51_B	38	24	1679	14	597	0.140726	2.17	0.9911	0.6095	0.3132	1.1861
H1_51_B:H7_51_B	28	24	1679	4	607	0.143502	2.14	1.0077	2.1691	0.7496	6.2772
H1_51_B:H8_51_B	21	20	1683	1	610	0.023788	5.11	1.0102	7.249	0.9708	54.1304
H1_51_B:H9_51_B	25	20	1683	5	606	0.466607	0.53	1.0036	1.4403	0.5382	3.8545
H1_51_B:H10_51_B	13	13	1690	0	611						
H1_51_B:H11_51_B	22	18	1685	4	607	0.380217	0.77	1.0041	1.6211	0.5465	4.809
H1_51_B:H12_51_B	12	10	1693	2	609						
H1_51_B:H13_51_B	13	13	1690	0	611						
H1_51_B:H14_51_B	14	7	1696	7	604						
H1_51_B:H15_51_B	13	8	1695	5	606						
H2_51_B:H2_51_B	53	47	1656	6	605	0.011738	6.35	1.0183	2.8618	1.2173	6.7278
H2_51_B:H3_51_B	50	39	1664	11	600	0.475139	0.51	1.005	1.2784	0.6505	2.5124
H2_51_B:H4_51_B	50	38	1665	12	599	0.698535	0.15	1.0027	1.1392	0.5914	2.1947
H2_51_B:H5_51_B	27	21	1682	6	605	0.617075	0.25	1.0025	1.2589	0.5057	3.134
H2_51_B:H6_51_B	26	23	1680	3	608	0.08378	2.99	1.0087	2.7746	0.8301	9.2741
H2_51_B:H7_51_B	18	15	1688	3	608						
H2_51_B:H8_51_B	12	10	1693	2	609						
H2_51_B:H9_51_B	10	10	1693	0	611						
H2_51_B:H10_51_B	14	14	1689	0	611						
H2_51_B:H11_51_B	10	8	1695	2	609						
H2_51_B:H12_51_B	10	7	1696	3	608						
H2_51_B:H13_51_B	10	8	1695	2	609						
H2_51_B:H14_51_B	10	7	1696	3	608						
H2_51_B:H15_51_B	9	7	1696	2	609						

FIG. 37

H3_51_B:H3_51_B	22	11	1692	11	600	0.011672	6.36	0.9884	0.3546	0.1529	0.8222
H3_51_B:H4_51_B	28	13	1690	15	596	0.001037	10.76	0.983	0.3056	0.1446	0.6461
H3_51_B:H5_51_B	22	11	1692	11	600	0.011672	6.36	0.9884	0.3546	0.1529	0.8222
H3_51_B:H6_51_B	11	6	1697	5	606			0.9953	0.4285	0.1303	1.4092
H3_51_B:H7_51_B	10	5	1698	5	606			0.9947	0.3569	0.103	1.2371
H3_51_B:H8_51_B	6	2	1701	4	607						
H3_51_B:H9_51_B	8	4	1699	4	607						
H3_51_B:H10_51_B	6	5	1698	1	610						
H4_51_B:H4_51_B	18	14	1689	4	607			1.0017	1.2578	0.4124	3.8362
H4_51_B:H5_51_B	18	9	1694	9	602			0.9905	0.3554	0.1404	0.8995
H4_51_B:H6_51_B	13	12	1691	1	610						
H4_51_B:H7_51_B	9	6	1697	3	608						
H4_51_B:H8_51_B	6	5	1698	1	610						
H4_51_B:H9_51_B	5	3	1700	2	609						
H4_51_B:H10_51_B	7	5	1698	2	609						
H5_51_B:H5_51_B	6	2	1701	4	607			0.9946	0.1784	0.0326	0.9766
H5_51_B:H6_51_B	13	8	1695	5	606			0.9965	0.572	0.1864	1.7553
H5_51_B:H7_51_B	8	6	1697	2	609						
H5_51_B:H8_51_B	7	4	1699	3	608						
H5_51_B:H9_51_B	4	3	1700	1	610						
H5_51_B:H10_51_B	0	0	1703	0	611						
H6_51_B:H6_51_B	3	1	1702	2	609						
H6_51_B:H7_51_B	3	3	1700	0	611						

FIG. 37 CONT.

Haplotype	Haplotype Sequence	rs35928059	rs3753394	rs551397	rs980292	rs16840419	rs3766404	rs16840422	rs1681147	rs1329421	rs10801554	rs1061170	rs12124794	rs695321	rs2274700	rs3753395	rs393955	rs3753396	rs403846	rs1410996	rs12144939	rs2284684	rs35292876	rs1065489	rs11562929
H1_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGGGGTTGCGGGCTTCGTCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H2_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGGGGTTGCGTATCTTCATCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H3_51_B	ACTAATCCTTTTCATAAGAGTCGCCGATACGGGGTTCCTCGTTTCGTCC	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	A	G	A	G	C	C	C
H4_51_B	ATCGATCCTTTACGAAGGGGCTTCGAATGCAAGTAGATTATCTTCATCC	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T
H5_51_B	ACCGGCTCTTTATATAAGATCCGCCGATGCAAGTCTGATCATCTTACTT	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	A	G	A	G	C	C	C
H6_51_B	ACTAATCCTTTTCATAAGAGTCGCCGATACGAAGTAGATTATCTTCATCC	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	A	G	A	G	C	C	C
H7_51_B	GTCGATCCTTTATGACAAGGCCGCCGACGCGGGTTCGTAICTTCATCC	G	T	C	G	A	T	C	C	T	T	T	A	T	G	A	C	A	A	G	G	C	C	G	C
H8_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGGAAGTAGATTATCTTCATCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H9_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTCAGGGTTCGCGGGCTTCGTCC	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T
H10_51_B	ATCGATCAACCATGACAAGGCCGCCGACGCGGGTTCGCGGGCTTCGTCC	A	T	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H11_51_B	ATCGATCCTTTACGAAGGGGCTTCGGATGCGGGTTCGCGGGCTTCGTCC	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T
H12_51_B	ATCGATCCTTTATATAAGATCCGCCGATGCAAGTAGATTATCTTCATCC	A	T	C	G	A	T	C	C	T	T	T	A	T	A	T	A	A	G	A	T	G	C	G	C
H13_51_B	ACCGATCAACCATGACAAGGCTGCCTGACGCGGGTTCGCGGGCTTCGTCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	T	G	C
H14_51_B	ACCGGCTCTTTATATAAGATCCGCCGATGCAAGTAGATTATCTTCATCC	A	C	C	G	C	T	C	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C
H15_51_B	ACCGGCTCTTTATATAAGATCCGCCGATGCAAGGTTGCGGGCTTCGTCC	A	C	C	G	C	T	C	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C
H16_51_B	ATCGGCTCTTTATATAAGATCCGCCGATGCAGGTCTGATCATCTTACTT	A	T	C	G	C	T	C	T	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C
H17_51_B	ACCGATCAACCATGACAAGGCCGCCGATACGGGTTGCTCGTTTCGTCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H18_51_B	ATCGATCCTTTACGAAGGGGCTTCGGATACGGGTTGCTCGTTTCGTCC	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	C	C	C	T	T
H19_51_B	ACCGATCAACCATGACAAGGCCGCCGATGCGGGTTCGCGGGCTTCGTCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H20_51_B	ACCGATCAACCATGACAAGGCCGCCGATGCAAGTAGATTATCTTCATCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H21_51_B	ATCGATCCTTTACGAAGGGGCTTCGGATGTGGGTTGCGGGCTTCGTCC	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	C	C	T	T	T
H22_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGGGTTCGCGGGCTTCGTCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H23_51_B	ATCGATCAACCATGACAAGGCCGCCGACGCGGGTTCGTAICTTCATCC	A	T	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H24_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGGGTTCGTAICTTCATCC	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H25_51_B	ATCGATCCTTTACGAAGGGGCTTCGAATGCAAGTAGATTATCTTCATCC	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	C	C	T	T	T
H26_51_B	ACTAATCCTTTACGAAGGGGCTTCGGATGTGGGTTGCGGGCTTCGTCC	A	C	T	A	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	C	C	T	T	T

FIG. 38-1

Haplotype	Haplotype Sequence	rs35928059	rs551397	rs800292	rs16840419	rs3766404	rs16840422	rs1061347	rs1329421	rs10801554	rs1061170	rs12124794	rs88985321	rs2274700	rs3753395	rs3753396	rs403846	rs1410996	rs12144939	rs2284664	rs35292876	rs1065489	rs11592399		
H27_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGGGGTTCGGGGCTTCGTCC	A	C	T	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	
H28_51_B	ATCGATCCTTTACGAAGGGGCTTCGGACGGGGGTTCGTAATCTTCATCC	A	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	
H29_51_B	GTCGATCAACCATGACAAGGCCCGCGGACGGGGGTTCGTAATCTTCATCC	G	T	C	G	A	T	C	A	A	C	A	T	G	A	C	A	G	A	T	C	C	G	C	
H30_51_B	ATCGATCCTTTATATAAGATCCGCCGGATACGGGGTTCGTAATCTTCATCC	A	T	C	G	A	T	C	C	T	T	T	T	A	T	A	A	G	A	T	C	C	G	C	
H31_51_B	ACCGATCAACCATGACAAGGCCCGCGGACGGGGGTTCGTAATCTTCATCC	A	C	G	A	T	C	A	A	C	A	C	T	G	A	C	A	G	A	T	C	C	G	C	
H32_51_B	ATCGATCCTTTATATAAGATCCGCCGAATGCAAGTAGATTATCTCCATCC	A	T	C	G	A	T	C	C	T	T	T	T	A	T	A	A	G	A	T	C	C	G	C	
H33_51_B	ACCGATCCTTTACGAAGGGGCTTCGAATGCAAGTAGATTATCTTCATCC	A	C	C	G	A	T	C	C	T	T	T	T	A	G	A	G	G	G	C	C	C	T	T	
H34_51_B	ATCGATCCTTTATATAAGATCCGCCGAATGCAAGTTCGGGGCTTCGTCC	A	T	C	G	A	T	C	C	T	T	T	T	A	T	A	A	G	A	T	C	C	G	C	
H35_51_B	GTCGATCAACCATGACAAGGCCCGCGGACGGGGGTTCGTAATCTTCATCC	G	T	C	G	A	T	C	A	A	C	A	T	G	A	C	A	G	G	C	C	C	G	C	
H36_51_B	ACCGATCAACCATGACAAGGCCCGCGGACGGGGGTTCGTAATCTTCATCC	A	C	C	G	A	T	C	A	A	C	A	T	G	A	C	A	G	G	C	C	C	G	C	
H37_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATCGGGGTTCGTAATCTTCATCC	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	G	A	G	T	C	G	C	
H38_51_B	ACTAATCCTTTACATAAGAGTCGCCGGATACGGGGTTCGTAATCTTCATCC	A	C	T	A	A	T	C	C	T	T	T	T	A	C	T	A	G	A	G	T	C	G	C	
H39_51_B	ACCGATCCTTTATGACAAGGCCCGCGGACGGGGGTTCGTAATCTTCATCC	A	C	C	G	A	T	C	C	T	T	T	T	A	G	A	C	A	G	G	C	C	G	C	
H40_51_B	ATCGGCTCTTTATATAAGATCCGCCGGATGCAAGGTTCGGGGCTTCGTCC	A	T	C	G	G	C	T	C	T	T	T	T	A	T	A	A	G	A	T	C	C	G	C	
H41_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGGGGTTCGTAATCTTCATCC	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C
H42_51_B	ACTAGCTCTTTATATAAGATCCGCCGAATGCAAGTAGATTATCTTCATCC	A	C	T	A	G	C	T	C	T	T	T	T	A	T	A	A	G	A	T	C	C	G	C	
H43_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGGGGTTCGTAATCTTCATCC	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C
H44_51_B	GCCGATCAACCATGACAAGGCCCGCGGACGGGGGTTCGTAATCTTCATCC	G	C	C	G	A	T	C	A	A	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C

FIG. 38-2

Haplotype	Haplotype Sequence	rs35928059	rs37533984	rs551397	rs800282	rs16840419	rs3766404	rs16840422	rs1061147	rs1929421	rs10801554	rs1061170	rs12124794	rs6095821	rs2274700	rs3753395	rs393955	rs3753396	rs403846	rs1410936	rs12144939	rs2284684	rs35292876	rs1065489	rs11582939
H45_51_B	ACCGATCCTTTTCATAAGAGTCGCCGGATACGGGGTTGCTCGTTTTCGTCC	A	C	C	G	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C
H46_51_B	ATOGATCCTTTACGAAGGGGCTTCGGATGTGGGTTGGGTATCTTCATCC	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T
H47_51_B	ATOGATCAACCATGACAAGGCTTCGAATGCAAGTAGATTATCTTCATCC	A	T	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	T	T
H48_51_B	ATOGATCCTTTATATAAGATCCTTCGAGTGC GG GTTGCGGGCTTCGTCC	A	T	C	G	A	T	C	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	T	T
H49_51_B	ACTAATCAACCATGACAAGGCGCGGACGGGGTTGGGGCTTCGTCC	A	C	T	A	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H50_51_B	ACCGATCAACCATGACAAGGCGCGGACGGGGCTGATCATCTTACTT	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H51_51_B	GTGGATCCTTTATGACAAGGCGCGGACGGGGTTGCTGTTTCGTCC	G	T	C	G	A	T	C	C	T	T	T	A	T	G	A	C	A	A	G	G	C	C	G	C
H52_51_B	GCCGATCAACCATGACAAGGCGCGGACGGGGTTGCGTATCTTCATCC	G	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C
H53_51_B	ACTAATCCTTTTCATAAGAGTCGCGGACGGGGTTGGGGCTTCGTCC	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C
H54_51_B	ACCGATCCTTTACGAAGGGGCTTCGGATGTGGGGTTGGGGCTTCGTCC	A	C	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T
H55_51_B	ATOGATCCTTTATATAAGATCGCGGAATGCAGGGTTGCGTATCTTCATCC	A	T	C	G	A	T	C	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C
H56_51_B	ATOGATCCTTTTCATAAGAGTCGCCGGATACGGGGTTGCTCGTTTTCGTCC	A	T	C	G	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C

FIG. 38-3

Haplotype	Haplotype Sequence	mrd_3876 (rs112797613)	mrd_3877	rs7546940	rs12405508 (mrd_3878)	rs12066959	mrd_3902 (rs79351096)	rs3828032	mrd_3905 (rs75816959)	rs6702340	rs3748557	rs12097550	rs11759311	rs10322138	rs598	rs2390516	rs10801586	rs5991	rs17514281	rs5897	rs1615413	rs6003	rs1412631 (mrd_3922)	rs6428380 (rs1115247)
H1_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGGGTTGCGGGCTTCGTCC	C	G	G	A	C	G	C	G	G	T	G	C	G	C	G	C	T	T	C	G	T	C	C
H2_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGGGTTGCGTATCTTCATCC	C	G	G	A	C	G	C	G	G	T	G	C	G	T	A	T	C	T	T	C	A	T	C
H3_51_B	ACTAATCCTTTTATAGAGTCGCCGATACGGGGTTGCTCGTTTCGTCC	C	G	G	A	T	A	C	G	G	T	G	C	T	G	T	G	T	T	T	G	T	C	C
H4_51_B	ATCGATCCTTTACGAAGGGGCTTCGAATGCAAAGTAGATTATCTTCATCC	C	G	A	A	T	G	C	A	A	G	A	G	A	T	A	T	A	C	T	T	A	T	C
H5_51_B	ACCGGCTCTTATATAAGATCCGCCGATGCAGGTCTGATCATCTTACTT	C	G	G	A	T	G	C	A	G	T	T	G	A	T	A	T	A	C	T	T	A	C	T
H6_51_B	ACTAATCCTTTTATAGAGTCGCCGATACGAAGTAGATTATCTTCATCC	C	G	G	A	T	A	C	G	A	G	T	A	G	A	T	A	T	A	C	T	G	A	T
H7_51_B	GTCGATCCTTTATGACAAGGCCGCCGACGCGGGTTGCGTATCTTCATCC	C	G	G	A	C	G	C	G	A	G	T	G	C	G	T	A	T	T	T	G	A	T	C
H8_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGAAGTAGATTATCTTCATCC	C	G	G	A	C	G	C	G	A	G	T	A	G	A	T	A	T	T	T	G	A	T	C
H9_51_B	ATCGATCCTTTACGAAGGGGCTTCGAGTGCAAGGTTGCGGGCTTCGTCC	C	G	A	G	T	G	C	A	G	G	T	T	G	C	G	C	G	T	T	G	C	T	C
H10_51_B	ATCGATCAACCATGACAAGGCCGCCGACGCGGGTTGCGGGCTTCGTCC	C	G	G	A	C	G	C	G	G	T	T	G	C	G	C	G	C	T	T	G	C	T	C
H11_51_B	ATCGATCCTTTACGAAGGGGCTTCGGAATGCGGGTTGCGGGCTTCGTCC	C	G	G	A	T	G	C	G	G	T	T	G	C	G	C	G	C	T	T	G	C	T	C
H12_51_B	ATCGATCCTTTATAGATCCGCCGATGCAAAGTAGATTATCTTCATCC	C	G	A	A	T	G	C	A	A	G	T	A	G	A	T	A	T	A	C	T	A	T	C
H13_51_B	ACCGATCAACCATGACAAGGCTGCCTGACGCGGGTTGCGGGCTTCGTCC	C	T	G	A	C	G	C	G	G	T	T	G	C	G	C	G	C	T	T	G	T	C	C
H14_51_B	ACCGGCTCTTATATAAGATCCGCCGATGCAAAGTAGATTATCTTCATCC	C	G	G	A	T	G	C	A	A	G	T	G	A	T	A	T	A	C	T	T	A	T	C
H15_51_B	ACCGGCTCTTATATAAGATCCGCCGATGCAGGTTGCGGGCTTCGTCC	C	G	G	A	T	G	C	A	G	G	T	T	G	C	G	C	G	C	T	T	G	T	C
H16_51_B	ATCGGCTCTTATATAAGATCCGCCGATGCAGGTCTGATCATCTTACTT	C	G	G	A	T	G	C	A	G	G	T	G	A	T	A	T	A	C	T	T	A	C	T
H17_51_B	ACCGATCAACCATGACAAGGCCGCCGATACGGGTTGCTCGTTTCGTCC	C	G	G	A	T	A	C	G	G	T	T	G	C	T	C	G	T	T	T	G	T	C	C
H18_51_B	ATCGATCCTTTACGAAGGGGCTTCGGAATACGGGTTGCTCGTTTCGTCC	C	G	G	A	T	A	C	G	G	T	T	G	C	T	C	G	T	T	T	G	T	C	C
H19_51_B	ACCGATCAACCATGACAAGGCCGCCGATGCGGGTTGCGGGCTTCGTCC	C	G	G	A	T	G	C	G	G	T	T	G	C	G	C	G	C	T	T	G	T	C	C
H20_51_B	ACCGATCAACCATGACAAGGCCGCCGATGCAAAGTAGATTATCTTCATCC	C	G	A	A	T	G	C	A	A	G	T	A	G	A	T	A	T	A	C	T	G	A	T
H21_51_B	ATCGATCCTTTACGAAGGGGCTTCGGAATGCGGGTTGCGGGCTTCGTCC	C	G	G	A	T	G	T	G	G	T	T	G	C	G	C	G	C	T	T	G	C	T	C
H22_51_B	ACCGATCAACCATGACAAGGCCGCCGACGCGGGTTACGGGCTTCGTCC	C	G	G	A	C	G	C	G	G	T	T	A	C	G	C	G	C	T	T	G	C	T	C
H23_51_B	ATCGATCAACCATGACAAGGCCGCCGATCCTTCATCC	C	G	G	A	C	G	C	G	G	T	T	G	C	G	T	A	T	C	T	T	A	T	C
H24_51_B	ACCGATCAACCATGACAAGGCCGCCGATGCGGGTTGCGTATCTTCATCC	G	G	G	A	C	G	C	G	G	T	T	G	C	G	T	A	T	C	T	T	A	T	C
H25_51_B	ATCGATCCTTTACGAAGGGGCTTCGAATGCAGGTTAGATTATCTTCATCC	C	G	A	A	T	G	C	A	G	G	T	A	G	A	T	A	T	C	T	T	A	T	C
H26_51_B	ACTAATCCTTTACGAAGGGGCTTCGGAATGCGGGTTGCGGGCTTCGTCC	C	G	G	A	T	G	T	G	G	T	T	G	C	G	C	G	C	T	T	G	T	C	C

FIG. 38-4

Haplotype	Haplotype Sequence	rs122797613	mrds_3877	rs7546940	rs12405503 (mrds_3879)	rs1498155	rs12066959	mrds_3902 (rs79351096)	rs9528032	mrds_3905 (rs75816959)	rs6702340	rs0437692	rs3748557	rs12097550	rs1770811	rs10822152	rs66839	rs3398	rs239510	rs10801586	rs5991	rs17514281	rs597	rs1615413	rs6003	rs1412631 (mrds_3922)	rs6428380 (rs1115247)
H27_51_B	ACTAATCCTTTTCATAAGAGTGCCTGCGGATACGGGGTTCGCGGCTTCGTCC	C	G	G	A	A	A	C	G	G	G	T	T	G	C	C	C	G	T	G	C	T	T	C	T	C	C
H28_51_B	ATCGATCCTTTACGAAGGGCTTCGACGCGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H29_51_B	GTCGATCAACCATGACAAGCGCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H30_51_B	ATCGATCCTTTATATAAGATCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H31_51_B	ACCGATCAACCATGACAAGCGCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H32_51_B	ATCGATCCTTTATATAAGATCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H33_51_B	ACCGATCCTTTACGAAGGGCTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H34_51_B	ATCGATCCTTTATATAAGATCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H35_51_B	GTCGATCAACCATGACAAGCGCGCGGATACGGGGTTCGATATCTTCATCC	G	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H36_51_B	ACCGATCAACCATGACAAGCGCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H37_51_B	ACTAATCCTTTTCATAAGAGTGCCTGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H38_51_B	ACTAATCCTTTTCATAAGAGTGCCTGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H39_51_B	ACCGATCCTTTATGACAAGCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H40_51_B	ATCGGCTCTTTATATAAGATCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H41_51_B	ACTAATCCTTTTCATAAGAGTGCCTGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H42_51_B	ACTAGCTCTTTATATAAGATCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H43_51_B	ACTAATCCTTTTCATAAGAGTGCCTGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C
H44_51_B	GCCGATCAACCATGACAAGCGCGCGGATACGGGGTTCGATATCTTCATCC	C	G	G	A	C	G	C	G	G	G	T	T	G	C	G	T	A	T	T	C	T	T	C	A	T	C

FIG. 38-5

Haplotype	Haplotype Sequence
H45_51_B	ACCGATCCTTTTCATAAGAGTCGCCGATACGGGGTTCCTCGTTTCGTCC
H46_51_B	ATCGATCCTTTACGAAGGGCCCTCGGATGTGGGGTTCGTTATCTTCATCC
H47_51_B	ATCGATCAACCATGACAAGGCCCTCGAATGCAAGTAGATTATCTTCATCC
H48_51_B	ATCGATCCTTTATATAAGATCCTTCGAGTGGGGGTTGCGGGCTTCGTCC
H49_51_B	ACTAATCAACCATGACAAGGCCGCCGACGCGGGGTTGCGGGCTTCGTCC
H50_51_B	ATCGATCAACCATGACAAGGCCGCCGACGCGGGTCTGATCTTTACTT
H51_51_B	GTCGATCCTTTATGACAAGGCCCGGATACGGGGTTCCTGTTTCGTCC
H52_51_B	GCCGATCAACCATGACAAGGCCGCCGACGCGGGTTCGTTATCTTCATCC
H53_51_B	ACTAATCCTTTTCATAAGATCGCCGACGCGGGGTTGCGGGCTTCGTCC
H54_51_B	ACCGATCCTTTACGAAGGGCCCTTCGGATGTGGGGTTCGCGGGCTTCGTCC
H55_51_B	ATCGATCCTTTATATAAGATCGCGGAATCGAGGGTTCGTTATCTTCATCC
H56_51_B	ATCGATCCTTTTCATAAGATCGCCGATACGGGGTTCCTCGTTTCGTCC

Haplotype	Freq.	Case, Control Ratio Counts	Case,Control Freqs	Chi Square	P Value
H1_51_B	0.214	1240.3 : 3884.9, 313.5 : 1480.7	0.242, 0.175	34.545	4.17E-09
H2_51_B	0.150	901.0 : 4224.2, 189.5 : 1604.7	0.176, 0.106	49.31	2.19E-12
H3_51_B	0.084	360.8 : 4764.4, 246.5 : 1547.6	0.070, 0.137	74.515	6.02E-18
H4_51_B	0.069	364.5 : 4760.7, 136.9 : 1657.2	0.071, 0.076	0.536	0.4642
H5_51_B	0.051	228.6 : 4896.5, 140.0 : 1654.2	0.045, 0.078	29.424	5.81E-08
H6_51_B	0.038	192.6 : 4932.6, 85.0 : 1709.2	0.038, 0.047	3.306	0.069
H7_51_B	0.028	147.9 : 4977.3, 52.4 : 1741.7	0.029, 0.029	0.006	0.9372
H8_51_B	0.02	107.7 : 5017.4, 35.0 : 1759.2	0.021, 0.019	0.155	0.6937
H9_51_B	0.018	101.8 : 5023.3, 31.9 : 1762.3	0.020, 0.018	0.311	0.5768
H10_51_B	0.018	104.0 : 5021.2, 27.2 : 1767.0	0.020, 0.015	1.888	0.1694
H11_51_B	0.017	94.7 : 5030.5, 28.0 : 1766.1	0.018, 0.016	0.614	0.4333
H12_51_B	0.017	67.0 : 5058.2, 55.6 : 1738.6	0.013, 0.031	24.48	7.51E-07
H13_51_B	0.015	91.4 : 5033.8, 16.0 : 1778.2	0.018, 0.009	6.911	0.0086
H14_51_B	0.013	48.6 : 5076.6, 46.9 : 1747.3	0.009, 0.026	27.149	1.88E-07
H15_51_B	0.012	53.4 : 5071.7, 32.7 : 1761.5	0.010, 0.018	6.563	0.0104
H16_51_B	0.011	54.2 : 5071.0, 28.8 : 1765.4	0.011, 0.016	3.362	0.0667
H17_51_B	0.011	70.5 : 5054.7, 11.3 : 1782.9	0.014, 0.006	6.348	0.0118
H18_51_B	0.011	59.8 : 5065.3, 17.9 : 1776.3	0.012, 0.010	0.355	0.5514
H19_51_B	0.010	54.3 : 5070.9, 17.2 : 1776.9	0.011, 0.010	0.126	0.7222
H20_51_B	0.010	47.9 : 5077.3, 23.4 : 1770.8	0.009, 0.013	1.78	0.1822
H21_51_B	0.010	49.6 : 5075.6, 19.8 : 1774.4	0.010, 0.011	0.236	0.6269
H22_51_B	0.010	55.9 : 5069.3, 13.0 : 1781.1	0.011, 0.007	1.782	0.1819
H23_51_B	0.009	54.0 : 5071.2, 10.4 : 1783.8	0.011, 0.006	3.264	0.0708
H24_51_B	0.008	50.0 : 5075.2, 8.0 : 1786.1	0.010, 0.004	4.426	0.0354
H25_51_B	0.006	31.7 : 5093.5, 14.4 : 1779.7	0.006, 0.008	0.696	0.4042
H26_51_B	0.006	28.9 : 5096.3, 16.0 : 1778.1	0.006, 0.009	2.246	0.134
H27_51_B	0.006	32.2 : 5092.9, 10.8 : 1783.3	0.006, 0.006	0.013	0.9099
H28_51_B	0.006	37.6 : 5087.6, 3.4 : 1790.7	0.007, 0.002	6.612	0.0101
H29_51_B	0.006	29.5 : 5095.7, 11.2 : 1783.0	0.006, 0.006	0.054	0.8164
H30_51_B	0.005	22.0 : 5103.2, 17.7 : 1776.5	0.004, 0.010	7.22	0.0072
H31_51_B	0.005	30.6 : 5094.6, 8.4 : 1785.8	0.006, 0.005	0.394	0.5301
H32_51_B	0.005	24.8 : 5100.4, 12.2 : 1782.0	0.005, 0.007	0.963	0.3264
H33_51_B	0.004	20.3 : 5104.8, 11.0 : 1783.2	0.004, 0.006	1.37	0.2418
H34_51_B	0.004	21.9 : 5103.3, 8.0 : 1786.2	0.004, 0.004	0.012	0.9112
H35_51_B	0.004	23.7 : 5101.4, 5.0 : 1789.2	0.005, 0.003	1.095	0.2953
H36_51_B	0.004	19.5 : 5105.6, 9.2 : 1785.0	0.004, 0.005	0.544	0.4607
H37_51_B	0.004	19.5 : 5105.6, 7.7 : 1786.5	0.004, 0.004	0.074	0.7857
H38_51_B	0.003	19.0 : 5106.2, 6.0 : 1788.2	0.004, 0.003	0.049	0.8254

FIG. 38-7

Haplotype	Freq.	Case, Control Ratio Counts	Case,Control Freqs	Chi Square	P Value
H39_51_B	0.003	18.8 : 5106.4, 6.0 : 1788.2	0.004, 0.003	0.045	0.8314
H40_51_B	0.003	12.0 : 5113.2, 9.1 : 1785.1	0.002, 0.005	3.216	0.0729
H41_51_B	0.003	16.0 : 5109.2, 4.8 : 1789.4	0.003, 0.003	0.091	0.7635
H42_51_B	0.003	13.0 : 5112.2, 6.0 : 1788.2	0.003, 0.003	0.316	0.5743
H43_51_B	0.002	8.1 : 5117.1, 8.9 : 1785.3	0.002, 0.005	6.221	0.0126
H44_51_B	0.002	13.3 : 5111.9, 2.0 : 1792.1	0.003, 0.001	1.287	0.2566
H45_51_B	0.002	9.4 : 5115.8, 4.0 : 1790.1	0.002, 0.002	0.118	0.7312
H46_51_B	0.002	7.2 : 5118.0, 4.2 : 1790.0	0.001, 0.002	0.72	0.3961
H47_51_B	0.001	7.2 : 5118.0, 3.0 : 1791.2	0.001, 0.002	0.059	0.8074
H48_51_B	0.001	10.0 : 5115.2, 0.0 : 1794.2	0.002, 0.000	3.506	0.0612
H49_51_B	0.001	6.7 : 5118.4, 3.2 : 1791.0	0.001, 0.002	0.194	0.6595
H50_51_B	0.001	7.1 : 5118.1, 2.2 : 1792.0	0.001, 0.001	0.024	0.8772
H51_51_B	0.001	6.2 : 5119.0, 2.7 : 1791.4	0.001, 0.002	0.101	0.7501
H52_51_B	0.001	7.8 : 5117.3, 1.0 : 1793.1	0.002, 0.001	0.938	0.3327
H53_51_B	0.001	5.9 : 5119.3, 1.5 : 1792.6	0.001, 0.001	0.105	0.7465
H54_51_B	0.001	5.4 : 5119.8, 2.0 : 1792.2	0.001, 0.001	0.003	0.9531
H55_51_B	0.001	4.3 : 5120.9, 3.1 : 1791.1	0.001, 0.002	0.946	0.3308
H56_51_B	0.001	4.8 : 5120.4, 2.5 : 1791.7	0.001, 0.001	0.247	0.6193

FIG. 38-8

FIG. 39

SNP Position on Figure 39	SNP	Position	MAF	Alleles	Relationship to Combined Cohort 51 SNP-based Haplotypes
001	rs10489457	194869137	0.000	C:C	
002	rs6695338	194870540	0.000	G:G	
003	rs12133754	194873556	0.000	C:C	
004	rs17573790	194876019	0.115	A:C	
005	rs16840355	194876583	0.000	A:A	
006	rs512822	194882461	0.013	G:T	
007	rs505102	194886125	0.218	T:C	
008	rs3753394	194887540	0.278	C:T	
009	rs7524776	194889960	0.141	T:C	Splits H3_51_B
010	rs12238983	194895889	0.000	C:C	
011	rs6680396	194899093	0.201	A:G	
012	rs16840401	194907880	0.004	A:G	
013	rs800292	194908856	0.218	G:A	
014	rs495222	194910006	0.004	C:T	
015	rs572515	194912884	0.368	G:A	
016	rs1061147	194920947	0.368	C:A	
017	rs7529589	194924902	0.368	C:T	
018	rs482934	194925120	0.372	T:G	
019	rs10801555	194926884	0.368	G:A	
020	rs10801556	194927087	0.368	G:A	
021	rs12405238	194928236	0.282	G:T	
022	rs10922096	194929082	0.470	T:C	
023	rs514943	194930536	0.368	T:C	
024	rs4658046	194937380	0.372	T:C	
025	rs10754199	194937462	0.368	G:A	
026	rs12038333	194939077	0.368	A:G	
027	rs1831282	194940616	0.415	C:A	
028	rs203687	194940893	0.415	T:C	
029	rs2019727	194941337	0.231	T:A	
030	rs2019724	194941540	0.415	C:T	
031	rs6428357	194942194	0.415	G:A	
032	rs7513157	194942303	0.030	G:A	Splits H5_51_B
033	rs6695321	194942484	0.359	A:G	
034	rs10733086	194943558	0.415	T:A	
035	rs1410997	194943786	0.415	A:C	
036	rs203684	194944632	0.000	C:C	
037	rs10737680	194946078	0.423	A:C	
038	rs16840465	194952599	0.000	A:A	

FIG. 40-1

SNP Position on Figure 39	SNP	Position	MAF	Alleles	Relationship to Combined Cohort 51 SNP-based Haplotypes
039	rs6677604	194953541	0.231	G:A	
040	rs379489	194960074	0.415	G:A	
041	rs419137	194963498	0.107	T:G	Splits H2_51_B
042	rs395544	194964895	0.415	G:A	
043	rs2284664	194969148	0.197	C:T	
044	rs1329428	194969433	0.423	C:T	
045	rs434536	194971790	0.004	C:T	
046	rs742855	194972143	0.162	T:C	
047	rs515299	194973300	0.000	G:G	
048	rs11798956	194973489	0.000	G:G	
049	rs543879	194973638	0.009	G:A	
050	rs424535	194975846	0.393	A:T	
051	rs11582939	194976780	0.162	C:T	
052	rs534399	194977690	0.000	G:G	
053	rs421820	195022923	0.158	C:T	
054	rs419436	195065612	0.026	C:T	
055	rs10922144	195089923	0.201	C:T	
056	rs16840658	195092251	0.111	G:A	
057	rs16840661	195092486	0.004	T:G	
058	rs6657442	195104683	0.209	T:C	
059	rs10737682	195108253	0.000	A:A	
060	rs7517836	195121926	0.000	C:C	
061	rs6428371	195122586	0.000	G:G	
062	rs12406509	195140815	0.038	A:G	
063	rs1409153	195146628	0.372	T:C	
064	rs7522952	195148967	0.120	A:G	
065	rs10494745	195154080	0.098	G:A	Splits H1_51_B and H10_51_B
066	rs7538501	195168376	0.154	C:T	
067	rs6674522	195182982	0.124	G:C	
068	rs9427649	195183076	0.000	G:G	
069	rs3828032	195186801	0.342	C:T	
070	rs7544166	195190963	0.000	A:A	
071	rs12723250	195196717	0.342	C:T	
072	rs9427938	195199868	0.124	C:T	
073	rs9427939	195203105	0.000	G:G	
074	rs6698935	195203678	0.000	A:A	
075	rs6428379	195204159	0.444	C:T	
076	rs6675769	195208284	0.487	G:A	
077	rs9427657	195209944	0.107	A:G	

FIG. 40-2

SNP Position on Figure 39	SNP	Position	MAF	Alleles	Relationship to Combined Cohort 51 SNP-based Haplotypes
078	rs6694672	195212412	0.085	T:G	
079	rs9427942	195213071	0.085	G:A	
080	rs9427661	195213169	0.085	T:C	
081	rs9427662	195213398	0.085	T:C	
082	rs12755054	195213653	0.303	T:C	
083	rs1170880	195213937	0.085	T:C	
084	rs1759016	195219121	0.303	C:T	
085	rs1750311	195220848	0.303	C:A	
086	rs16840943	195228576	0.000	G:G	
087	rs12092294	195233476	0.085	A:G	
088	rs12116643	195239806	0.162	T:C	
089	rs16840956	195244312	0.000	T:T	
090	rs10922153	195245238	0.466	G:T	
091	rs6663083	195247283	0.466	C:T	
092	rs7555070	195248777	0.303	T:C	
093	rs1332666	195251302	0.466	G:A	
094	rs2336597	195259962	0.085	G:A	
095	rs1360558	195260596	0.385	G:A	
096	rs2336596	195261715	0.470	A:G	
097	rs7418993	195266670	0.000	G:G	
098	rs1332668	195272359	0.470	T:C	
099	rs1332669	195272506	0.085	T:C	
100	rs13375369	195273852	0.004	G:A	
101	rs857021	195274102	0.085	G:A	
102	rs698859	195274988	0.385	C:T	
103	rs5898	195276421	0.470	G:A	
104	rs10754209	195278200	0.470	A:T	
105	rs7518773	195278259	0.470	G:A	
106	rs10754210	195278734	0.380	G:A	
107	rs6000	195286481	0.000	A:A	
108	rs6001	195286560	0.000	T:T	
109	rs17549671	195286602	0.000	A:A	
110	rs2990510	195287281	0.380	T:G	
111	rs5899	195292888	0.000	T:T	
112	rs17514281	195292912	0.009	A:G	
113	rs1759009	195293295	0.090	G:A	
114	rs1759007	195294229	0.090	G:A	
115	rs17514253	195296710	0.017	C:T	

FIG. 40-3

SNP Position on Figure 39	SNP	Position	MAF	Alleles	Relationship to Combined Cohort 51 SNP-based Haplotypes
116	rs5997	195296824	0.090	C:T	
117	rs1627765	195299666	0.094	A:G	
118	rs4915148	195302161	0.090	C:T	
119	rs1412632	195303491	0.090	A:G	

FIG. 40-4

FIG.
41-1

SEQ ID NO.	Reference Haplotype No.	Haplotypes	rs3753394	rs800292	rs106147	rs6695321	rs2284664	rs11582939	rs12406509 (mrd_3878)	rs1409153	rs3828032	rs9427662	rs1750311	rs10922153	rs698859	rs5998	rs2990510	rs17514281	rs5997
288	H1_51_B	CGATCCACGTCGGGGTC	C	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
289	H2_51_B	CGATCCACGTCGGGGTC	C	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
290	H3_51_B	CACCTCAATGTCGGTC	C	A	C	C	T	C	A	T	G	T	C	T	C	G	T	T	C
291	H4_51_B	TGCCCTAATATATATTC	T	G	C	C	C	T	A	T	A	T	A	T	T	A	T	T	C
292	H5_51_B	CGCTCCATACATCATTC	C	G	C	T	C	C	A	T	A	C	A	T	C	A	T	T	T
293	H6_51_B	CACCTCAATGTCGGTC	C	A	C	C	T	C	A	T	G	T	A	T	T	A	T	T	C
294	H7_51_B	TGCTCCACGTCGGGGTC	T	G	C	T	C	C	A	C	G	T	C	G	T	A	T	T	C
295	H8_51_B	CGATCCACGTCGGGGTC	C	G	A	T	C	C	A	C	G	T	A	T	T	A	T	T	C
296	H9_51_B	TGCCCTGTCGGGGTC	T	G	C	C	C	T	G	T	A	T	C	G	C	G	G	T	C
297	H10_51_B	TGATCCACGTCGGGGTC	T	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
298	H11_51_B	TGCCCTAATGTCGGGGTC	T	G	C	C	C	T	A	T	G	T	C	G	C	G	G	T	C
299	H12_51_B	TGCTCCATATATATTC	T	G	C	T	C	C	A	T	A	T	A	T	T	A	T	T	C
300	H13_51_B	CGATCCACGTCGGGGTC	C	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
301	H14_51_B	CGCTCCATATATATTC	C	G	C	T	C	C	A	T	A	T	A	T	T	A	T	T	C
302	H15_51_B	CGCTCCATATGCGGGTC	C	G	C	T	C	C	A	T	A	T	C	G	C	G	G	T	C
303	H16_51_B	TGCTCCATACATCATTC	T	G	C	T	C	C	A	T	A	C	A	T	C	A	T	T	T
304	H17_51_B	CGATCCATGTCGGGGTC	C	G	A	T	C	C	A	T	G	T	C	T	C	G	T	T	C
305	H18_51_B	TGCCCTAATGTCGGTC	T	G	C	C	C	T	A	T	G	T	C	T	C	G	T	T	C
306	H19_51_B	CGATCCATGTCGGGGTC	C	G	A	T	C	C	A	T	G	T	C	G	C	G	G	T	C
307	H20_51_B	CGATCCATATATATTC	C	G	A	T	C	C	A	T	A	T	A	T	T	A	T	T	C
308	H21_51_B	TGCCCTAATGTCGGGGTC	T	G	C	C	C	T	A	T	G	T	C	G	C	G	G	T	C
309	H22_51_B	CGATCCACGTCGGGGTC	C	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
310	H23_51_B	TGATCCACGTCGGTC	T	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
311	H24_51_B	CGATCCACGTCGGTC	C	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
312	H25_51_B	TGCCCTAATATATATTC	T	G	C	C	C	T	A	T	A	T	A	T	T	A	T	T	C

FIG. 41-2

SEQ. ID NO.	Reference Haplotype No.	Haplotypes	rs37533594	rs800292	rs1061147	rs6695321	rs2284664	rs11582939	rs12406509 (chr1_3878)	rs1409153	rs3828032	rs9427662	rs1750311	rs10922153	rs698839	rs5998	rs2990510	rs17514281	rs5997
313	H26_51_B	CACCTCATGTCGGGTC	C	A	C	C	C	T	A	T	G	T	C	G	C	G	G	T	C
314	H27_51_B	CACCTCATGTCGGGTC	C	A	C	C	T	C	A	T	G	T	C	G	C	G	G	T	C
315	H28_51_B	TGCCCCAGTCGTATTC	T	G	C	C	C	T	A	C	G	T	C	G	T	A	T	T	C
316	H29_51_B	TGATCCACGTCGTATTC	T	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
317	H30_51_B	TGCTCCATGTCGTTC	T	G	C	T	C	C	A	T	G	T	C	T	C	G	T	T	C
318	H31_51_B	CGATCCACGTCGGGTC	C	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
319	H32_51_B	TGCTCCATATTAATCC	T	G	C	T	C	C	A	T	A	T	A	T	T	A	T	C	C
320	H33_51_B	CGCCCCATATTAATTC	C	G	C	C	C	T	A	T	A	T	A	T	T	A	T	T	C
321	H34_51_B	TGCTCCATATCGGGTC	T	G	C	T	C	C	A	T	A	T	C	G	C	G	G	T	C
322	H35_51_B	TGATCCACGTCGTATTC	T	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
323	H36_51_B	CGATCCACGTCGTTC	C	G	A	T	C	C	A	C	G	T	C	T	C	G	T	T	C
324	H37_51_B	CACCTCATGTCGTATTC	C	A	C	C	T	C	A	T	G	T	C	G	T	A	T	T	C
325	H38_51_B	CACCTCATGTCGTTC	C	A	C	C	T	C	A	T	G	T	C	T	C	G	T	T	C
326	H39_51_B	CGCTCCACGTCGTATTC	C	G	C	T	C	C	A	C	G	T	C	G	T	A	T	T	C
327	H40_51_B	TGCTCCATATCGGGTC	T	G	C	T	C	C	A	T	A	T	C	G	C	G	G	T	C
328	H41_51_B	CACCTCATGTCGTATTC	C	A	C	C	T	C	A	T	G	T	C	G	T	A	T	T	C
329	H42_51_B	CACCTCATATTAATTC	C	A	C	T	C	C	A	T	A	T	A	T	T	A	T	T	C
330	H43_51_B	CACCTCATGTCGGTC	C	A	C	C	T	C	A	T	G	T	C	T	C	G	G	T	C
331	H44_51_B	CGATCCACGTCGGGTC	C	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
332	H45_51_B	CGCTCATGTCGTTC	C	G	C	C	T	C	A	T	G	T	C	T	C	G	T	T	C
333	H46_51_B	TGCCCTATGTCGTATTC	T	G	C	C	C	T	A	T	G	T	C	G	T	A	T	T	C
334	H47_51_B	TGATCTATATTAATTC	T	G	A	T	C	T	A	T	A	T	A	T	T	A	T	T	C
335	H48_51_B	TGCTCTGTGTCGGGTC	T	G	C	T	C	T	G	T	G	T	C	G	C	G	G	T	C
336	H49_51_B	CAATCCACGTCGGGTC	C	A	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
337	H50_51_B	CGATCCACGCATCATTT	C	G	A	T	C	C	A	C	G	C	A	T	C	A	T	T	T

FIG.
41-3

SEQ ID NO.	Reference Haplotype No.	Haplotypes	rs3753394	rs800292	rs1061147	rs6695321	rs2284664	rs11582939	rs12406509 (mtd_3878)	rs1409153	rs3828032	rs9427662	rs1750311	rs10922153	rs698859	rs5998	rs2990510	rs17514281	rs5997
338	H51_51_B	TGCTCCATGTCGTTTC	T	G	C	T	C	C	A	T	G	T	C	T	C	G	T	T	C
339	H52_51_B	CGATCCACGTGGTATTC	C	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
340	H53_51_B	CACCTCACGTGGGGTC	C	A	C	C	T	C	A	C	G	T	C	G	C	G	G	T	C
341	H54_51_B	CGCCCTATGTCGGGGTC	C	G	C	C	C	T	A	T	G	T	C	G	C	G	G	T	C
342	H55_51_B	TGCTCCATATCGTATTC	T	G	C	T	C	C	A	T	A	T	C	G	T	A	T	T	C
343	H56_51_B	TGCCTCATGTCGTTTC	T	G	C	C	T	C	A	T	G	T	C	T	C	G	T	T	C

FIG.
41-4

SEQ ID NO.	51-SNP, Combined Cohort Haplotypes Tagged	HAPMAP Haplotypes	rs3753394	rs800292	rs1061147	rs6695321	rs2284664	rs11582939	rs12406509 (mrd_3878)	rs1409153	rs3828032	rs9427662	rs1750311	rs10922153	rs698859	rs5998	rs2990510	rs17514281	rs5997
344	1,13,22,31,44	OGATCCAGTGGGTC	C	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
345	1,13,22,31,44	OGATCCAGTGGGTC	C	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
346	3,38	CACTCATGTCTGTTTC	C	A	C	C	T	C	A	T	G	T	C	T	C	G	T	T	C
347	4,25	TGOCCTATATATTTC	T	G	C	C	C	T	A	T	A	T	A	T	T	A	T	T	C
348	2,24,52	CGATCCACGTGGTATTC	C	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
349	6	CACCTCATGTATTATTC	C	A	C	C	T	C	A	T	G	T	A	T	T	A	T	T	C
350	5	CGCTCCATACATCATTT	C	G	C	T	C	C	A	T	A	C	A	T	C	A	T	T	T
351	3,38	CACTCATGTCTGTTTC	C	A	C	C	T	C	A	T	G	T	C	T	C	G	T	T	C
352	7	TGCTCCAGTGGTATTC	T	G	C	T	C	C	A	C	G	T	C	G	T	A	T	T	C
353	9	TGOCCTGTATGGGTC	T	G	C	C	C	T	G	T	A	T	C	G	C	G	G	T	C
354	11,21	TGOCCTATGTGGGTC	T	G	C	C	C	T	A	T	G	T	C	G	C	G	G	T	C
355	2,24,52	CGATCCACGTGGTATTC	C	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
356	5	CGCTCCATACATCATTT	C	G	C	T	C	C	A	T	A	C	A	T	C	A	T	T	T
357	12	TGCTCCATATATTATTC	T	G	C	T	C	C	A	T	A	T	A	T	T	A	T	T	C
358	14	CGCTCCATATATTATTC	C	G	C	T	C	C	A	T	A	T	A	T	T	A	T	T	C
359	8	CGATCCAGTATTATTC	C	G	A	T	C	C	A	C	G	T	A	T	T	A	T	T	C
360	34,40	TGCTCCATATGGGTC	T	G	C	T	C	C	A	T	A	T	C	G	C	G	G	T	C
361	15	CGCTCCATATGGGTC	C	G	C	T	C	C	A	T	A	T	C	G	C	G	G	T	C
362	2,24,52	CGATCCACGTGGTATTC	C	G	A	T	C	C	A	C	G	T	C	G	T	A	T	T	C
363	20	OGATCCATATATTATTC	C	G	A	T	C	C	A	T	A	T	A	T	T	A	T	T	C
364	19	CGATCCATGTGGGTC	C	G	A	T	C	C	A	T	G	T	C	G	C	G	G	T	C
365	43	CACCTCATGTCTGGTC	C	A	C	C	T	C	A	T	G	T	C	T	C	G	G	T	C

**FIG.
41-5**

SEQ ID NO.	51-SNP, Combined Cohort Haplotypes Tagged	HAPMAP Haplotypes	rs3753394	rs800292	rs1061147	rs6695321	rs2284664	rs11582939	rs12406509 (mrd_3878)	rs1409153	rs3828032	rs9427662	rs1750311	rs10922153	rs698859	rs5998	rs2990510	rs17514281	rs5997
366	17	CGATCCATGTCTCGTTC	C	G	A	T	C	C	A	T	G	T	C	T	C	G	T	T	C
367	17	CGATCCATGTCTCGTTC	C	G	A	T	C	C	A	T	G	T	C	T	C	G	T	T	C
368	37,41	CACCTCATGTGTTATTC	C	A	C	C	T	C	A	T	G	T	C	G	T	A	T	T	C
369	39	CGCTCCACGTGTTATTC	C	G	C	T	C	C	A	C	G	T	C	G	T	A	T	T	C
370	4,25	TGOCCTATATTTATTC	T	G	C	C	C	T	A	T	A	T	A	T	T	A	T	T	C
371	15	CGCTCCATATGCGGGTC	C	G	C	T	C	C	A	T	A	T	C	G	C	G	G	T	C
372	10	TGATCCACGTGCGGGTC	T	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
373	10	TGATCCACGTGCGGGTC	T	G	A	T	C	C	A	C	G	T	C	G	C	G	G	T	C
374	N/A	CGCTCCATGTGCGGGTC	C	G	C	T	C	C	A	T	G	T	C	G	C	G	G	T	C
375	32	TGCTCCATATTTATTC	T	G	C	T	C	C	A	T	A	T	A	T	T	A	T	C	C
376	5	CGCTCCATACATCATTT	C	G	C	T	C	C	A	T	A	C	A	T	C	A	T	T	T

SNP No.	Name	Assoc Allele	Case, Control Ratio Counts	Case,Control Freqs	Chi square	P value
1	rs35928059	G	263:5229, 84:1824	0.048, 0.044	0.473	0.4917
2	rs3753394	C	4003:1465, 1379:543	0.732, 0.717	1.531	0.216
3	rs551397	C	4696:796, 1468:442	0.855, 0.769	76.088	2.71E-18
4	rs800292	G	4667:793, 1466:444	0.855, 0.768	77.074	1.65E-18
5	rs16840419	A	5049:435, 1621:283	0.921, 0.851	77.387	1.41E-18
6	rs3766404	T	4980:456, 1612:290	0.916, 0.848	72.574	1.61E-17
7	rs16840422	C	5010:460, 1618:292	0.916, 0.847	73.191	1.18E-17
8	rs1061147	A	3043:2431, 735:1185	0.556, 0.383	170.424	5.98E-39
9	rs1329421	A	3054:2434, 732:1174	0.556, 0.384	168.356	1.69E-38
10	rs10801554	C	2997:2369, 718:1144	0.559, 0.386	165.444	7.32E-38
11	rs1061170	C	3045:2427, 730:1174	0.556, 0.383	169.323	1.04E-38
12	rs12124794	A	4770:712, 1511:401	0.870, 0.790	70.683	4.19E-17
13	rs6695321	T	3855:1607, 1200:712	0.706, 0.628	40.143	2.36E-10
14	rs2274700	G	4130:1326, 1113:793	0.757, 0.584	206.287	8.87E-47
15	rs3753395	A	4139:1331, 1116:792	0.757, 0.585	203.639	3.36E-46
16	rs393955	C	3228:2244, 795:1111	0.590, 0.417	170.253	6.51E-39
17	rs3753396	G	896:4586, 313:1607	0.163, 0.163	0.002	0.9656
18	rs403846	A	3213:2229, 796:1102	0.590, 0.419	166.046	5.40E-38
19	rs1410996	G	4111:1329, 1109:797	0.756, 0.582	207.442	4.97E-47
20	rs12144939	G	4885:609, 1514:396	0.889, 0.793	112.46	2.83E-26
21	rs2284664	C	4757:725, 1515:399	0.868, 0.792	63.939	1.28E-15
22	rs35292876	T	95:5387, 15:1907	0.017, 0.008	8.821	0.003
23	rs1065489	G	4567:909, 1600:322	0.834, 0.832	0.024	0.8763
24	rs11582939	C	4552:906, 1595:321	0.834, 0.832	0.024	0.8761
25	mrd__3876	G	78:5404, 13:1905	0.014, 0.007	6.494	0.0108
26	mrd__3877	T	89:5333, 18:1880	0.016, 0.009	4.689	0.0304
27	rs7546940	G	4639:799, 1561:333	0.853, 0.824	8.98	0.0027
28	rs12406509	G	131:5337, 37:1883	0.024, 0.019	1.405	0.236
29	rs1409153	C	3055:2359, 746:1150	0.564, 0.393	164.157	1.40E-37
30	rs12066959	G	4592:876, 1450:444	0.840, 0.766	52.661	3.96E-13
31	mrd__3902	C	5341:95, 1870:42	0.983, 0.978	1.559	0.2118
32	rs3828032	G	4255:1209, 1296:612	0.779, 0.679	75.255	4.14E-18
33	mrd__3905	G	4461:999, 1459:459	0.817, 0.761	28.419	9.77E-08
34	mrd__3906	G	4463:1005, 1456:460	0.816, 0.760	28.265	1.06E-07
35	rs6702340	G	5158:316, 1721:191	0.942, 0.900	39.412	3.43E-10
36	rs9427662	T	5132:312, 1726:192	0.943, 0.900	40.728	1.75E-10
37	rs3748557	T	4451:1013, 1446:470	0.815, 0.755	31.707	1.79E-08
38	rs12097550	A	62:5420, 13:1903	0.011, 0.007	2.897	0.0888
39	rs1750311	C	4128:1338, 1247:659	0.755, 0.654	72.939	1.34E-17
40	rs10922153	G	3508:1954, 918:994	0.642, 0.480	155.143	1.30E-35

FIG. 42-1

SNP No.	Name	Assoc Allele	Case, Control Ratio Counts	Case,Control Freqs	Chi square	P value
41	rs698859	T	2408:3058, 785:1129	0.441, 0.410	5.339	0.0209
42	rs5998	G	2743:2729, 940:976	0.501, 0.491	0.647	0.4213
43	rs2990510	G	2131:3339, 613:1311	0.390, 0.319	30.72	2.98E-08
44	rs10801586	C	4863:611, 1589:329	0.888, 0.828	45.936	1.22E-11
45	rs5991	A	32:5434, 10:1904	0.006, 0.005	0.099	0.7526
46	rs17514281	T	5388:32, 1889:13	0.994, 0.993	0.2	0.6549
47	rs5997	C	5142:322, 1720:188	0.941, 0.901	34.441	4.39E-09
48	rs1615413	G	2732:2736, 931:979	0.500, 0.487	0.843	0.3586
49	rs6003	T	5135:321, 1710:192	0.941, 0.899	38.563	5.30E-10
50	rs1412631	C	5144:322, 1718:186	0.941, 0.902	33.09	8.80E-09
51	rs6428380	C	5138:324, 1720:190	0.941, 0.901	35.184	3.00E-09

FIG. 42-2

SNP No.	Name	Assoc Allele	Case, Control Ratio Counts	Case, Control Freqs	Chi square	P value
1	rs35928059	A	1830:82, 1824:84	0.957, 0.956	0.03	0.863
2	rs3753394	C	1417:485, 1379:543	0.745, 0.717	3.684	0.0549
3	rs551397	C	1573:341, 1468:442	0.822, 0.769	16.649	4.50E-05
4	rs800292	G	1553:339, 1466:444	0.821, 0.768	16.503	4.86E-05
5	rs16840419	A	1713:199, 1621:283	0.896, 0.851	17.161	3.43E-05
6	rs3766404	T	1684:208, 1612:290	0.890, 0.848	15.049	1.00E-04
7	rs16840422	C	1704:208, 1618:292	0.891, 0.847	16.337	5.30E-05
8	rs1061147	A	963:949, 735:1185	0.504, 0.383	56.698	5.08E-14
9	rs1329421	A	965:947, 732:1174	0.505, 0.384	56.277	6.30E-14
10	rs10801554	C	954:934, 718:1144	0.505, 0.386	54.356	1.67E-13
11	rs1061170	C	960:944, 730:1174	0.504, 0.383	56.278	6.29E-14
12	rs12124794	A	1598:312, 1511:401	0.837, 0.790	13.543	2.00E-04
13	rs6695321	T	1310:594, 1200:712	0.688, 0.628	15.466	8.40E-05
14	rs2274700	G	1314:584, 1113:793	0.692, 0.584	48.352	3.56E-12
15	rs3753395	A	1325:585, 1116:792	0.694, 0.585	49.011	2.54E-12
16	rs393955	C	1029:877, 795:1111	0.540, 0.417	57.563	3.27E-14
17	rs3753396	A	1631:285, 1607:313	0.851, 0.837	1.485	0.223
18	rs403846	A	1028:876, 796:1102	0.540, 0.419	55.321	1.02E-13
19	rs1410996	G	1307:583, 1109:797	0.692, 0.582	49.346	2.15E-12
20	rs12144939	G	1643:269, 1514:396	0.859, 0.793	29.524	5.52E-08
21	rs2284664	C	1595:315, 1515:399	0.835, 0.792	11.936	6.00E-04
22	rs35292876	T	23:1887, 15:1907	0.012, 0.008	1.752	0.1856
23	rs1065489	G	1625:287, 1600:322	0.850, 0.832	2.179	0.1399
24	rs11582939	C	1623:285, 1595:321	0.851, 0.832	2.366	0.124
25	mrd_3876	G	16:1896, 13:1905	0.008, 0.007	0.322	0.5703
26	mrd_3877	T	19:1877, 18:1880	0.010, 0.009	0.028	0.8662
27	rs7546940	G	1625:277, 1561:333	0.854, 0.824	6.41	0.0113
28	rs12406509	G	46:1860, 37:1883	0.024, 0.019	1.066	0.3018
29	rs1409153	C	975:917, 746:1150	0.515, 0.393	56.732	5.00E-14
30	rs12066959	G	1543:365, 1450:444	0.809, 0.766	10.553	0.0012
31	mrd_3902	C	1866:30, 1870:42	0.984, 0.978	1.937	0.164

FIG. 43-1

SNP No.	Name	Assoc Allele	Case, Control Ratio Counts	Case, Control Freqs	Chi square	P value
32	rs3828032	G	1443:465, 1296:612	0.756, 0.679	27.953	1.24E-07
33	mrsl_3905	G	1535:367, 1459:459	0.807, 0.761	12.109	5.00E-04
34	mrsl_3906	G	1533:371, 1456:460	0.805, 0.760	11.478	7.00E-04
35	rs6702340	G	1766:142, 1721:191	0.926, 0.900	7.787	0.0053
36	rs9427662	T	1761:141, 1726:192	0.926, 0.900	8.095	0.0044
37	rs3748557	T	1537:365, 1446:470	0.808, 0.755	15.929	6.58E-05
38	rs12097550	A	15:1897, 13:1903	0.008, 0.007	0.148	0.7003
39	rs1750311	C	1390:510, 1247:659	0.732, 0.654	26.737	2.33E-07
40	rs10922153	G	1129:771, 918:994	0.594, 0.480	49.887	1.63E-12
41	rs698859	T	825:1075, 785:1129	0.434, 0.410	2.265	0.1323
42	rs5998	A	973:935, 976:940	0.510, 0.509	0.001	0.9722
43	rs2990510	G	678:1228, 613:1311	0.356, 0.319	5.901	0.0151
44	rs10801586	C	1645:257, 1589:329	0.865, 0.828	9.749	0.0018
45	rs5991	A	14:1886, 10:1904	0.007, 0.005	0.701	0.4025
46	rs17514281	C	20:1870, 13:1889	0.011, 0.007	1.543	0.2142
47	rs5997	C	1765:143, 1720:188	0.925, 0.901	6.699	0.0096
48	rs1615413	G	930:976, 931:979	0.488, 0.487	0.001	0.9754
49	rs6003	T	1763:143, 1710:192	0.925, 0.899	7.972	0.0048
50	rs1412631	C	1765:143, 1718:186	0.925, 0.902	6.25	0.0124
51	rs6428380	C	1751:143, 1720:190	0.924, 0.901	6.843	0.0089

FIG. 43-2

SNP No.	Name	Assoc Allele	Case, Control Ratio Counts	Case, Control Freqs	Chi square	P value
1	rs35928059	G	30:474, 84:1824	0.060, 0.044	2.127	0.1447
2	rs3753394	C	364:140, 1379:543	0.722, 0.717	0.044	0.8332
3	rs551397	C	451:53, 1468:442	0.895, 0.769	38.996	4.25E-10
4	rs890292	G	452:52, 1466:444	0.897, 0.768	40.83	1.66E-10
5	rs16840419	A	478:28, 1621:283	0.945, 0.851	30.961	2.63E-08
6	rs3766404	T	476:28, 1612:290	0.944, 0.848	32.626	1.12E-08
7	rs16840422	C	473:31, 1618:292	0.938, 0.847	28.725	8.34E-08
8	rs1061147	A	299:199, 735:1185	0.600, 0.383	76.49	2.21E-18
9	rs1329421	A	304:202, 732:1174	0.601, 0.384	76.657	2.03E-18
10	rs10801554	C	299:189, 718:1144	0.613, 0.386	81.235	2.08E-19
11	rs1061170	C	304:202, 730:1174	0.601, 0.383	77.119	1.61E-18
12	rs12124794	A	459:47, 1511:401	0.907, 0.790	36.187	1.79E-09
13	rs6695321	T	359:139, 1200:712	0.721, 0.628	15.046	1.00E-04
14	rs2274700	G	407:97, 1113:793	0.808, 0.584	85.558	2.25E-20
15	rs3753395	A	404:96, 1116:792	0.808, 0.585	84.708	3.46E-20
16	rs393955	C	314:190, 795:1111	0.623, 0.417	68.034	1.61E-16
17	rs3753396	G	92:410, 313:1607	0.183, 0.163	1.171	0.2791
18	rs403846	A	313:189, 796:1102	0.624, 0.419	66.545	3.42E-16
19	rs1410996	G	406:98, 1109:797	0.806, 0.582	85.448	2.38E-20
20	rs12144939	G	457:49, 1514:396	0.903, 0.793	32.501	1.19E-08
21	rs2284664	C	456:48, 1515:399	0.905, 0.792	33.941	5.68E-09
22	rs35292876	T	9:497, 15:1907	0.018, 0.008	4.078	0.0434
23	rs1065489	T	95:407, 322:1600	0.189, 0.168	1.317	0.2511
24	rs11582939	T	95:407, 321:1595	0.189, 0.168	1.316	0.2514
25	mrd_3876	G	8:498, 13:1905	0.016, 0.007	3.803	0.0512
26	mrd_3877	T	8:494, 18:1880	0.016, 0.009	1.542	0.2142
27	rs7546940	G	416:80, 1561:333	0.839, 0.824	0.58	0.4462
28	rs12406509	G	11:491, 37:1883	0.022, 0.019	0.143	0.7054
29	rs1409153	C	292:200, 746:1150	0.593, 0.393	63.61	1.52E-15
30	rs12066959	G	431:75, 1450:444	0.852, 0.766	17.508	2.86E-05
31	mrd_3902	C	491:9, 1870:42	0.982, 0.978	0.301	0.5831

FIG. 44-1

SNP No.	Name	Assoc Allele	Case, Control Ratio Counts	Case, Control Freqs	Chi square	P value
32	rs3828032	G	389:111, 1296:612	0.778, 0.679	18.39	1.80E-05
33	mrd_3905	G	413:89, 1459:459	0.823, 0.761	8.737	0.0031
34	mrd_3906	G	416:88, 1456:460	0.825, 0.760	9.767	0.0018
35	rs6702340	G	478:24, 1721:191	0.952, 0.900	13.296	3.00E-04
36	rs9427662	T	475:23, 1726:192	0.954, 0.900	14.178	2.00E-04
37	rs3748557	T	414:90, 1446:470	0.821, 0.755	9.991	0.0016
38	rs12097550	A	6:500, 13:1903	0.012, 0.007	1.323	0.25
39	rs1750311	C	387:115, 1247:659	0.771, 0.654	24.796	6.37E-07
40	rs10922153	G	330:172, 918:994	0.657, 0.480	50.022	1.52E-12
41	rs698859	T	221:281, 785:1129	0.440, 0.410	1.483	0.2233
42	rs5998	G	258:244, 940:976	0.514, 0.491	0.867	0.3519
43	rs2990510	G	202:302, 613:1311	0.401, 0.319	12.097	5.00E-04
44	rs10801586	C	446:56, 1589:329	0.888, 0.828	10.699	0.0011
45	rs5991	T	502:2, 1904:10	0.996, 0.995	0.128	0.721
46	rs17514281	T	496:0, 1889:13	1.000, 0.993	3.409	0.0649
47	rs5997	C	476:24, 1720:188	0.952, 0.901	12.6	4.00E-04
48	rs1615413	G	257:245, 931:979	0.512, 0.487	0.956	0.3282
49	rs6003	T	480:24, 1710:192	0.952, 0.899	13.866	2.00E-04
50	rs1412631	C	481:25, 1718:186	0.951, 0.902	11.665	6.00E-04
51	rs6428380	C	481:25, 1720:190	0.951, 0.901	12.37	4.00E-04

FIG. 44-2

SNP No.	Name	Assoc Allele	Case, Control Ratio Counts	Case, Control Freqs	Chi square	P value
1	rs35928059	G	130:2624, 84:1824	0.047, 0.044	0.26	0.6101
2	rs3753394	C	1983:759, 1379:543	0.723, 0.717	0.183	0.6686
3	rs551397	C	2378:374, 1468:442	0.864, 0.769	71.232	3.18E-17
4	rs800292	G	2370:374, 1466:444	0.864, 0.768	71.884	2.28E-17
5	rs16840419	A	2559:187, 1621:283	0.932, 0.851	80.264	3.28E-19
6	rs3766404	T	2522:198, 1612:290	0.927, 0.848	75.246	4.16E-18
7	rs16840422	C	2535:199, 1618:292	0.927, 0.847	76.289	2.45E-18
8	rs1061147	A	1579:1163, 735:1185	0.576, 0.383	168.343	1.70E-38
9	rs1329421	A	1583:1165, 732:1174	0.576, 0.384	165.962	5.64E-38
10	rs10801554	C	1545:1129, 718:1144	0.578, 0.386	162.16	3.82E-37
11	rs1061170	C	1579:1161, 730:1174	0.576, 0.383	167.165	3.08E-38
12	rs12124794	A	2418:326, 1511:401	0.881, 0.790	70.7	4.16E-17
13	rs6695321	T	1942:796, 1200:712	0.709, 0.628	34.261	4.82E-09
14	rs2274700	G	2144:588, 1113:793	0.785, 0.584	216.556	5.10E-49
15	rs3753395	A	2146:594, 1116:792	0.783, 0.585	211.358	6.94E-48
16	rs393955	C	1670:1070, 795:1111	0.609, 0.417	167.041	3.28E-38
17	rs3753396	G	469:2277, 313:1607	0.171, 0.163	0.489	0.4842
18	rs403846	A	1657:1057, 796:1102	0.611, 0.419	163.904	1.59E-37
19	rs1410996	G	2135:591, 1109:797	0.783, 0.582	216.7	4.74E-49
20	rs12144939	G	2494:262, 1514:396	0.905, 0.793	117.379	2.37E-27
21	rs2284664	C	2412:334, 1515:399	0.878, 0.792	64.155	1.15E-15
22	rs35292876	T	57:2689, 15:1907	0.021, 0.008	12.492	4.00E-04
23	rs1065489	T	476:2264, 322:1600	0.174, 0.168	0.305	0.5808
24	rs11582939	T	476:2252, 321:1595	0.174, 0.168	0.382	0.5363
25	mrd_3876	G	49:2693, 13:1905	0.018, 0.007	10.577	0.0011
26	mrd_3877	T	55:2647, 18:1880	0.020, 0.009	8.437	0.0037
27	rs7546940	G	2323:399, 1561:333	0.853, 0.824	7.154	0.0075
28	rs12406509	G	64:2674, 37:1883	0.023, 0.019	0.896	0.3438
29	rs1409153	C	1582:1126, 746:1150	0.584, 0.393	162.304	3.55E-37
30	rs12066959	G	2332:404, 1450:444	0.852, 0.766	56.316	6.17E-14
31	mrd_3902	C	2669:51, 1870:42	0.981, 0.978	0.59	0.4423

FIG. 45-1

SNP No.	Name	Assoc Allele	Case, Control Ratio Counts	Case, Control Freqs	Chi square	P value
32	rs3828032	G	2168:568, 1296:612	0.792, 0.679	75.937	2.93E-18
33	mrd_3905	G	2252:484, 1459:459	0.823, 0.761	27.184	1.85E-07
34	mrd_3906	G	2253:487, 1456:460	0.822, 0.760	27.05	1.98E-07
35	rs6702340	G	2612:130, 1721:191	0.953, 0.900	48.323	3.61E-12
36	rs9427662	T	2594:128, 1726:192	0.953, 0.900	49.372	2.12E-12
37	rs3748557	T	2239:497, 1446:470	0.818, 0.755	27.726	1.40E-07
38	rs12097550	A	38:2706, 13:1903	0.014, 0.007	5.2	0.0226
39	rs1750311	C	2110:632, 1247:659	0.770, 0.654	74.464	6.17E-18
40	rs10922153	G	1828:910, 918:994	0.668, 0.480	163.712	1.75E-37
41	rs698859	T	1204:1540, 785:1129	0.439, 0.410	3.78	0.0519
42	rs5998	G	1407:1333, 940:976	0.514, 0.491	2.365	0.1241
43	rs2990510	G	1128:1612, 613:1311	0.412, 0.319	41.853	9.84E-11
44	rs10801586	C	2470:278, 1589:329	0.899, 0.828	49.426	2.06E-12
45	rs5991	T	2726:14, 1904:10	0.995, 0.995	0.003	0.957
46	rs17514281	T	2701:11, 1889:13	0.996, 0.993	1.668	0.1965
47	rs5997	C	2599:135, 1720:188	0.951, 0.901	41.939	9.42E-11
48	rs1615413	G	1403:1335, 931:979	0.512, 0.487	2.809	0.0937
49	rs6003	T	2594:134, 1710:192	0.951, 0.899	45.988	1.19E-11
50	rs1412631	C	2598:134, 1718:186	0.951, 0.902	41.31	1.30E-10
51	rs6428380	C	2604:136, 1720:190	0.950, 0.901	42.886	5.80E-11

FIG. 45-2

Haplotype	Controls (Stage 0) vs Geographis Atrophy (Stage 4A)
H1_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGCGGCTTCGTCC
H2_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGTATCTTCATCC
H3_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGGGGTTGCTCGTTTTCGTCC
H4_51_B	ATCGATCCTTTACGAAGGGGGCCTTCGAATGCAAAGTAGATTATCTTCATCC
H5_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAGGTCTGATCATCTTTACTT
H6_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGAAGTAGATTATCTTCATCC
H7_51_B	GTCGATCCTTTATGACAAGGCCGCCGGACGCGGGGTTGCGTATCTTCATCC
H8_51_B	ATCGATCCTTTATATAAGATCCGCCGAATGCAAAGTAGATTATCTTCATCC
H9_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAAAGTAGATTATCTTCATCC
H10_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGAAGTAGATTATCTTCATCC
H11_51_B	ATCGATCCTTTACGAAGGGGGCCTTCGAGTGCAGGGTTGCGCGGCTTCGTCC
H12_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAGGGTTGCGCGGCTTCGTCC
H13_51_B	ATCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGCGGCTTCGTCC
H14_51_B	ATCGGCTCTTTATATAAGATCCGCCGGATGCAGGTCTGATCATCTTTACTT
H15_51_B	ACCGATCAACCATGACAAGGCCGCCGAATGCAAAGTAGATTATCTTCATCC
H16_51_B	ATCGATCCTTTACGAAGGGGGCCTTCGGATGCGGGGTTGCGCGGCTTCGTCC
H17_51_B	ATCGATCCTTTACGAAGGGGGCCTTCGGATACGGGGTTGCTCGTTTTCGTCC
H18_51_B	ACCGATCAACCATGACAAGGCCGCCGGATGCGGGGTTGCGCGGCTTCGTCC

Haplotype	Controls (Stage 0) vs CNV (Stage 4B)
H1_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGCGGCTTCGTCC
H2_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGTATCTTCATCC
H3_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGGGGTTGCTCGTTTTCGTCC
H4_51_B	ATCGATCCTTTACGAAGGGGGCCTTCGAATGCAAAGTAGATTATCTTCATCC
H5_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAGGTCTGATCATCTTTACTT
H6_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGAAGTAGATTATCTTCATCC
H7_51_B	GTCGATCCTTTATGACAAGGCCGCCGGACGCGGGGTTGCGTATCTTCATCC
H8_51_B	ATCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGCGGCTTCGTCC
H9_51_B	ATCGATCCTTTACGAAGGGGGCCTTCGAGTGCAGGGTTGCGCGGCTTCGTCC
H10_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGAAGTAGATTATCTTCATCC
H11_51_B	ATCGATCCTTTATATAAGATCCGCCGAATGCAAAGTAGATTATCTTCATCC

FIG. 46-1

Haplotype	Controls (Stage 0) vs CNV (Stage 4B)
H12_51_B	ATCGATCCTTTACGAAGGGGCCCTTCGGATGCGGGGTTGCGCGGCTTCGTCC
H13_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAAAGTAGATTATCTTCATCC
H14_51_B	ACCGATCAACCATGACAAGGCTGCCTGACGCGGGGTTGCGCGGCTTCGTCC
H15_51_B	ATCGGCTCTTTATATAAGATCCGCCGGATGCAGGTCTGATCATCTTTACTT
H16_51_B	ACCGATCAACCATGACAAGGCCGCCGGATGCGGGGTTGCGCGGCTTCGTCC
H17_51_B	ATCGATCCTTTACGAAGGGGCCCTTCGGATGTGGGGTTGCGCGGCTTCGTCC
H18_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAGGGTTGCGCGGCTTCGTCC
H19_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTACGCGGCTTCGTCC
H20_51_B	ATCGATCCTTTACGAAGGGGCCCTTCGGATACGGGGTTGCTCGTTTTTCGTCC
H21_51_B	ACCGATCAACCATGACAAGGCCGCCGAATGCAAAGTAGATTATCTTCATCC

Haplotype	Controls (Stage 0) vs Early Stage AMD (Stages 1B-3)
H1_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGCGGCTTCGTCC
H2_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGTATCTTCATCC
H3_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGGGGTTGCTCGTTTTTCGTCC
H4_51_B	ATCGATCCTTTACGAAGGGGCCCTTCGAATGCAAAGTAGATTATCTTCATCC
H5_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAGGTCTGATCATCTTTACTT
H6_51_B	ACTAATCCTTTTCATAAGAGTCGCCGGATACGAAGTAGATTATCTTCATCC
H7_51_B	GTCGATCCTTTATGACAAGGCCGCCGGACGCGGGGTTGCGTATCTTCATCC
H8_51_B	ATCGATCCTTTATATAAGATCCGCCGAATGCAAAGTAGATTATCTTCATCC
H9_51_B	ACCGATCAACCATGACAAGGCCGCCGGACGCGAAGTAGATTATCTTCATCC
H10_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAAAGTAGATTATCTTCATCC
H11_51_B	ATCGATCCTTTACGAAGGGGCCCTTCGAGTGCAGGGTTGCGCGGCTTCGTCC
H12_51_B	ACCGGCTCTTTATATAAGATCCGCCGGATGCAGGGTTGCGCGGCTTCGTCC
H13_51_B	ATCGATCCTTTACGAAGGGGCCCTTCGGATGCGGGGTTGCGCGGCTTCGTCC
H14_51_B	ATCGATCAACCATGACAAGGCCGCCGGACGCGGGGTTGCGCGGCTTCGTCC
H15_51_B	ATCGGCTCTTTATATAAGATCCGCCGGATGCAGGTCTGATCATCTTTACTT
H16_51_B	ACCGATCAACCATGACAAGGCCGCCGAATGCAAAGTAGATTATCTTCATCC

FIG. 46-2

	Controls (Stage 0) vs Geographis Atrophy (Stage 4A)																									
Haplotype	rs35928059	rs3753394	rs551397	rs5800292	rs16840419	rs3766404	rs16840422	rs1061147	rs1329421	rs10801554	rs1061170	rs12124794	rs6695321	rs2274700	rs3753395	rs393955	rs3753396	rs403846	rs1410996	rs12144939	rs2284664	rs35292876	rs1065489	rs11582939	mrd_3876 (rs112797613)	mrd_3877
H1_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H2_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H3_S1_B	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G
H4_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H5_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H6_S1_B	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G
H7_S1_B	G	T	C	G	A	T	C	C	T	T	T	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H8_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H9_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H10_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H11_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H12_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H13_S1_B	A	T	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H14_S1_B	A	T	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H15_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H16_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H17_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H18_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G

FIG. 46-3

	Controls (Stage 0) vs CNV (Stage 4B)																									
Haplotype	rs35928059	rs3753394	rs551397	rs800292	rs16840419	rs3766404	rs16840422	rs1061147	rs13329421	rs10801554	rs1061170	rs12124794	rs6695321	rs2274700	rs3753395	rs393955	rs3753396	rs403846	rs1410996	rs12144939	rs2284664	rs35292876	rs1065489	rs11582939	mrd_3876 (rs112797613)	mrd_3877
H1_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H2_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H3_S1_B	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G
H4_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H5_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H6_S1_B	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G
H7_S1_B	G	T	C	G	A	T	C	C	T	T	T	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H8_S1_B	A	T	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H9_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H10_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H11_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H12_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H13_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H14_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	T	G	C	C	T
H15_S1_B	A	T	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H16_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H17_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H18_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H19_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H20_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H21_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G

FIG. 46-4

	Controls (Stage 0) vs Early Stage AMD (Stages 1B-3)																									
Haplotype	rs35928059	rs3753394	rs551397	rs800292	rs16840419	rs3766404	rs16840422	rs1061147	rs1329421	rs10801554	rs1061170	rs12124794	rs6695321	rs3274700	rs3753395	rs393955	rs3753396	rs403846	rs1410996	rs12144939	rs2284664	rs35292876	rs1065489	rs11582939	mrd_3876 (rs112797613)	mrd_3877
H1_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H2_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H3_S1_B	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G
H4_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H5_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H6_S1_B	A	C	T	A	A	T	C	C	T	T	T	T	C	A	T	A	A	G	A	G	T	C	G	C	C	G
H7_S1_B	G	T	C	G	A	T	C	C	T	T	T	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H8_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H9_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H10_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H11_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H12_S1_B	A	C	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H13_S1_B	A	T	C	G	A	T	C	C	T	T	T	A	C	G	A	A	G	G	G	G	C	C	T	T	C	G
H14_S1_B	A	T	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G
H15_S1_B	A	T	C	G	G	C	T	C	T	T	T	A	T	A	T	A	A	G	A	T	C	C	G	C	C	G
H16_S1_B	A	C	C	G	A	T	C	A	A	C	C	A	T	G	A	C	A	A	G	G	C	C	G	C	C	G

FIG. 46-5

	Controls (Stage 0) vs Geographis Atrophy (Stage 4A)																														
Haplotype	rs7546940	rs12406509 (mrd_3878)		rs1409153	rs12066959		mrd_3902 (rs79351096)		rs3828032	mrd_3905 (rs75816959)		mrd_3906 (rs74188363)		rs6702340	rs9427662	rs3748557	rs12097550	rs1750311	rs10922153	rs698859	rs5998	rs2990510	rs10801586	rs5991	rs17514281	rs5997	rs1615413	rs6003	rs1412631 (mrd_3922)	rs6428380 (rs1115247)	
H1_51_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	G	C	G	G	C	T	T	C	G	T	C	C	
H2_51_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	T	A	T	C	T	T	C	A	T	C	A	T	C	A	T	C	C
H3_51_B	G	A	T	A	C	G	G	G	G	T	T	G	C	T	C	G	T	T	T	T	C	G	T	C	C	T	C	G	T	C	C
H4_51_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T	C	A	T	C	A	T	C	A	T	C	C
H5_51_B	G	A	T	G	C	A	G	G	T	C	T	G	A	T	C	A	T	C	T	T	T	A	C	T	T	A	C	T	T	T	T
H6_51_B	G	A	T	A	C	G	A	A	G	T	A	G	A	T	T	A	T	C	T	T	C	A	T	C	A	T	C	A	T	C	C
H7_51_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	T	A	T	C	G	T	A	T	C	T	T	C	A	T	C	C	C
H8_51_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T	C	A	T	C	A	T	C	A	T	C	C
H9_51_B	G	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T	C	A	T	C	A	T	C	A	T	C	C
H10_51_B	G	A	C	G	C	G	A	A	G	T	A	G	A	T	T	A	T	C	T	T	C	A	T	C	A	T	C	A	T	C	C
H11_51_B	A	G	T	G	C	A	G	G	G	T	T	G	C	G	C	G	G	C	T	T	C	G	T	C	G	T	C	G	T	C	C
H12_51_B	G	A	T	G	C	A	G	G	G	T	T	G	C	G	C	G	G	C	T	T	C	G	T	C	G	T	C	G	T	C	C
H13_51_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T	C	G	T	C	G	T	C	G	T	C	C
H14_51_B	G	A	T	G	C	A	G	G	T	C	T	G	A	T	C	A	T	C	T	T	T	A	C	T	T	A	C	T	T	T	T
H15_51_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T	C	A	T	C	A	T	C	A	T	C	C
H16_51_B	G	A	T	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T	C	G	T	C	G	T	C	G	T	C	C
H17_51_B	G	A	T	A	C	G	G	G	G	T	T	G	C	T	C	G	T	T	T	T	C	G	T	C	G	T	C	G	T	C	C
H18_51_B	G	A	T	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T	C	G	T	C	G	T	C	G	T	C	C

FIG. 46-6

	Controls (Stage 0) vs CNV (Stage 4B)																			
Haplotype	rs7546940	rs12406509 (mrd_3878)	rs1409153	rs12066959	mrd_3902 (rs79351096)	rs3828032	mrd_3905 (rs75816959)	mrd_3906 (rs74188363)	rs6702340	rs9427662	rs3748557	rs12097550	rs1750311	rs10922153	rs698859	rs5998	rs2990510	rs10801586	rs5991	rs17514281
H1_S1_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H2_S1_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	T	A	T	C	T	T
H3_S1_B	G	A	T	A	C	G	G	G	G	T	T	G	C	T	C	G	T	T	T	T
H4_S1_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H5_S1_B	G	A	T	G	C	A	G	G	T	C	T	G	A	T	C	A	T	C	T	T
H6_S1_B	G	A	T	A	C	G	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H7_S1_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	T	A	T	C	T	T
H8_S1_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H9_S1_B	A	G	T	G	C	A	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H10_S1_B	G	A	C	G	C	G	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H11_S1_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H12_S1_B	G	A	T	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H13_S1_B	G	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H14_S1_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H15_S1_B	G	A	T	G	C	A	G	G	T	C	T	G	A	T	C	A	T	C	T	T
H16_S1_B	G	A	T	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H17_S1_B	G	A	T	G	T	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H18_S1_B	G	A	T	G	C	A	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H19_S1_B	G	A	C	G	C	G	G	G	G	T	T	A	C	G	C	G	G	C	T	T
H20_S1_B	G	A	T	A	C	G	G	G	G	T	T	G	C	T	C	G	T	T	T	T
H21_S1_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T

FIG. 46-7

	Controls (Stage 0) vs Early Stage AMD (Stages 1B-3)																			
Haplotype	rs7546940	rs12406509 (mrd_3878)	rs1409153	rs12066959	mrd_3902 (rs79351096)	rs3828032	mrd_3905 (rs75816959)	mrd_3906 (rs74188363)	rs6702340	rs9427662	rs3748557	rs12097550	rs1750311	rs10922153	rs698859	rs5998	rs2990510	rs10801586	rs5991	rs17514281
H1_51_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H2_51_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	T	A	T	C	T	T
H3_51_B	G	A	T	A	C	G	G	G	G	T	T	G	C	T	C	G	T	T	T	T
H4_51_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H5_51_B	G	A	T	G	C	A	G	G	T	C	T	G	A	T	C	A	T	C	T	T
H6_51_B	G	A	T	A	C	G	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H7_51_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	T	A	T	C	T	T
H8_51_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H9_51_B	G	A	C	G	C	G	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H10_51_B	G	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T
H11_51_B	A	G	T	G	C	A	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H12_51_B	G	A	T	G	C	A	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H13_51_B	G	A	T	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H14_51_B	G	A	C	G	C	G	G	G	G	T	T	G	C	G	C	G	G	C	T	T
H15_51_B	G	A	T	G	C	A	G	G	T	C	T	G	A	T	C	A	T	C	T	T
H16_51_B	A	A	T	G	C	A	A	A	G	T	A	G	A	T	T	A	T	C	T	T

FIG. 46-8

Controls (Stage 0) vs Geographis Atrophy (Stage 4A)					
Haplotype	Freq.	Case, Control Ratio Counts	Case, Control Freqs	Chi Square	P Value
H1_S1_B	0.182	118.7 : 356.2, 312.1 : 1487.9	0.250, 0.173	14.336	2.00E-04
H2_S1_B	0.117	90.1 : 384.8, 186.6 : 1613.4	0.190, 0.104	26.081	3.27E-07
H3_S1_B	0.113	25.0 : 449.9, 243.7 : 1556.3	0.053, 0.135	24.74	6.56E-07
H4_S1_B	0.075	40.6 : 434.2, 137.9 : 1662.1	0.086, 0.077	0.418	0.518
H5_S1_B	0.067	19.5 : 455.4, 138.8 : 1661.2	0.041, 0.077	7.545	0.006
H6_S1_B	0.041	11.9 : 463.0, 85.9 : 1714.1	0.025, 0.048	4.69	0.0303
H7_S1_B	0.028	11.0 : 463.9, 54.3 : 1745.7	0.023, 0.030	0.663	0.4156
H8_S1_B	0.026	8.3 : 466.6, 53.3 : 1746.7	0.017, 0.030	2.103	0.147
H9_S1_B	0.02	0.0 : 474.9, 46.5 : 1753.5	0.000, 0.026	12.496	4.00E-04
H10_S1_B	0.02	11.1 : 463.8, 35.3 : 1764.7	0.023, 0.020	0.259	0.6106
H11_S1_B	0.016	7.2 : 467.7, 31.3 : 1768.7	0.015, 0.017	0.117	0.7323
H12_S1_B	0.015	4.9 : 470.0, 31.6 : 1768.4	0.010, 0.018	1.281	0.2577
H13_S1_B	0.015	8.0 : 466.9, 27.4 : 1772.6	0.017, 0.015	0.057	0.8109
H14_S1_B	0.014	1.9 : 473.0, 32.1 : 1767.9	0.004, 0.018	4.927	0.0264
H15_S1_B	0.014	6.4 : 468.5, 26.5 : 1773.5	0.013, 0.015	0.047	0.8292
H16_S1_B	0.014	5.1 : 469.8, 27.7 : 1772.3	0.011, 0.015	0.601	0.4384
H17_S1_B	0.011	9.7 : 465.2, 17.2 : 1782.8	0.020, 0.010	3.78	0.0519
H18_S1_B	0.01	6.8 : 468.1, 17.7 : 1782.3	0.014, 0.010	0.701	0.4026

Controls (Stage 0) vs CNV (Stage 4B)					
Haplotype	Freq.	Case, Control Ratio Counts	Case, Control Freqs	Chi Square	P Value
H1_S1_B	0.209	648.1 : 1947.4, 310.5 : 1483.8	0.250, 0.173	36.541	1.49E-09
H2_S1_B	0.141	461.8 : 2133.7, 186.7 : 1607.6	0.178, 0.104	45.992	1.19E-11
H3_S1_B	0.087	158.3 : 2437.2, 242.7 : 1551.6	0.061, 0.135	70.504	4.59E-17
H4_S1_B	0.07	185.6 : 2409.9, 137.6 : 1656.7	0.072, 0.077	0.414	0.5199
H5_S1_B	0.05	91.3 : 2504.2, 138.2 : 1656.1	0.035, 0.077	37.489	9.19E-10
H6_S1_B	0.038	91.0 : 2504.5, 85.4 : 1708.8	0.035, 0.048	4.329	0.0375
H7_S1_B	0.028	73.6 : 2521.9, 54.8 : 1739.5	0.028, 0.031	0.178	0.6731
H8_S1_B	0.02	66.4 : 2529.1, 27.6 : 1766.6	0.026, 0.015	5.249	0.022
H9_S1_B	0.018	53.0 : 2542.5, 31.9 : 1762.4	0.020, 0.018	0.388	0.5333
H10_S1_B	0.018	49.9 : 2545.6, 34.9 : 1759.4	0.019, 0.019	0.002	0.9625

FIG. 46-9

Controls (Stage 0) vs CNV (Stage 4B)					
Haplotype	Freq.	Case, Control Ratio Counts	Case, Control Freqs	Chi Square	P Value
H11_51_B	0.018	28.7 : 2566.8, 52.9 : 1741.3	0.011, 0.030	19.788	8.65E-06
H12_51_B	0.017	49.3 : 2546.2, 28.0 : 1766.3	0.019, 0.016	0.708	0.4001
H13_51_B	0.016	24.4 : 2571.1, 47.4 : 1746.8	0.009, 0.026	19.162	1.20E-05
H14_51_B	0.016	55.5 : 2540.0, 16.0 : 1778.3	0.021, 0.009	10.278	0.0013
H15_51_B	0.014	30.6 : 2564.9, 32.6 : 1761.6	0.012, 0.018	3.057	0.0804
H16_51_B	0.012	36.6 : 2558.9, 17.9 : 1776.3	0.014, 0.010	1.47	0.2253
H17_51_B	0.011	32.0 : 2563.5, 19.5 : 1774.7	0.012, 0.011	0.19	0.663
H18_51_B	0.01	16.9 : 2578.6, 31.2 : 1763.0	0.006, 0.017	11.648	6.00E-04
H19_51_B	0.01	34.2 : 2561.3, 13.0 : 1781.2	0.013, 0.007	3.485	0.0619
H20_51_B	0.01	29.7 : 2565.8, 17.4 : 1776.9	0.011, 0.010	0.305	0.581
H21_51_B	0.01	21.8 : 2573.7, 24.2 : 1770.0	0.008, 0.013	2.632	0.1047

Controls (Stage 0) vs Early Stage AMD (Stages 1B-3)					
Haplotype	Freq.	Case, Control Ratio Counts	Case, Control Freqs	Chi Square	P Value
H1_51_B	0.192	404.4 : 1390.1, 313.2 : 1490.3	0.225, 0.174	15.061	1.00E-04
H2_51_B	0.125	281.7 : 1512.9, 184.5 : 1619.0	0.157, 0.102	23.849	1.04E-06
H3_51_B	0.111	168.1 : 1626.4, 245.9 : 1557.5	0.094, 0.136	16.088	6.05E-05
H4_51_B	0.068	112.2 : 1682.3, 140.6 : 1662.8	0.063, 0.078	3.272	0.0705
H5_51_B	0.065	103.8 : 1690.8, 138.3 : 1665.1	0.058, 0.077	5.093	0.024
H6_51_B	0.044	78.6 : 1715.9, 84.8 : 1718.6	0.044, 0.047	0.216	0.6422
H7_51_B	0.03	60.3 : 1734.2, 52.5 : 1750.9	0.034, 0.029	0.599	0.439
H8_51_B	0.022	28.8 : 1765.7, 55.2 : 1748.3	0.016, 0.031	8.312	0.0039
H9_51_B	0.02	39.5 : 1755.0, 34.9 : 1768.5	0.022, 0.019	0.31	0.5774
H10_51_B	0.019	23.6 : 1770.9, 46.8 : 1756.7	0.013, 0.026	7.627	0.0057
H11_51_B	0.018	35.7 : 1758.8, 31.9 : 1771.6	0.020, 0.018	0.242	0.6229
H12_51_B	0.017	29.3 : 1766.3, 33.4 : 1770.1	0.016, 0.019	0.254	0.6145
H13_51_B	0.017	33.8 : 1760.7, 28.2 : 1775.3	0.019, 0.016	0.558	0.4551
H14_51_B	0.014	26.3 : 1768.3, 26.9 : 1776.6	0.015, 0.015	0.004	0.9489
H15_51_B	0.012	18.2 : 1776.4, 28.5 : 1775.0	0.010, 0.016	2.236	0.1348
H16_51_B	0.012	19.6 : 1775.0, 23.8 : 1779.7	0.011, 0.013	0.392	0.5314

FIG. 46-10

Diplotype	Cases with	Cases w/o	Controls with	Controls w/o	p-value
H1_51_B:H1_51_B	42	813	26	1047	0.003253
H1_51_B:H2_51_B	70	785	46	1027	0.000347
H1_51_B:H3_51_B	18	837	49	1024	0.00338
H1_51_B:H4_51_B	21	834	25	1048	0.86249
H1_51_B:H5_51_B	12	843	32	1041	0.021082
H1_51_B:H6_51_B	13	842	18	1055	0.791337
H1_51_B:H7_51_B	11	844	7	1066	0.150222
H1_51_B:H8_51_B	11	844	5	1068	0.048574
H1_51_B:H9_51_B	12	843	8	1065	0.156265
H1_51_B:H10_51_B	7	848	4	1069	
H1_51_B:H11_51_B	4	851	6	1067	
H1_51_B:H12_51_B	8	847	5	1068	
H1_51_B:H13_51_B	3	852	1	1072	
H1_51_B:H14_51_B	3	852	14	1059	0.026091
H2_51_B:H2_51_B	22	833	7	1066	0.000577
H2_51_B:H3_51_B	18	837	19	1054	0.596701
H2_51_B:H4_51_B	10	845	19	1054	0.281466
H2_51_B:H5_51_B	15	840	15	1058	0.532299
H2_51_B:H6_51_B	8	847	7	1066	
H2_51_B:H7_51_B	9	846	7	1066	
H2_51_B:H8_51_B	3	852	5	1068	
H2_51_B:H9_51_B	5	850	3	1070	
H2_51_B:H10_51_B	4	851	1	1072	
H3_51_B:H3_51_B	13	842	19	1054	0.671373
H3_51_B:H4_51_B	12	843	25	1048	0.140726
H3_51_B:H5_51_B	11	844	14	1059	1
H3_51_B:H6_51_B	3	852	11	1062	0.083265
H3_51_B:H7_51_B	8	847	7	1066	0.483927
H3_51_B:H8_51_B	1	854	5	1068	
H3_51_B:H9_51_B	0	855	4	1069	
H4_51_B:H4_51_B	6	849	5	1068	
H4_51_B:H5_51_B	3	852	12	1061	0.056747
H4_51_B:H6_51_B	4	851	1	1072	
H4_51_B:H7_51_B	1	854	4	1069	
H5_51_B:H5_51_B	3	852	5	1068	
H5_51_B:H6_51_B	4	851	9	1064	
H5_51_B:H7_51_B	3	852	4	1069	

FIG. 47-1

Diplotype	Chi Square (Pearson)	Risk Ratio	Odds Ratio	95% CI lower	95% CI upper
H1_S1_B:H1_S1_B	18.73	1.0387	2.598	1.661	4.0636
H1_S1_B:H2_S1_B	15.42	1.0429	2.0009	1.4071	2.8454
H1_S1_B:H3_S1_B	3.6	0.9849	0.6691	0.4407	1.0159
H1_S1_B:H4_S1_B	0.41	1.0042	1.1817	0.7088	1.9702
H1_S1_B:H5_S1_B	5.48	0.9858	0.5247	0.3031	0.9083
H1_S1_B:H6_S1_B	0.22	0.9976	0.8581	0.4517	1.6304
H1_S1_B:H7_S1_B	0.91	1.0036	1.5539	0.625	3.8637
H1_S1_B:H8_S1_B	3.39	1.007	2.4946	0.911	6.8313
H1_S1_B:H9_S1_B					
H1_S1_B:H10_S1_B					
H1_S1_B:H11_S1_B					
H1_S1_B:H12_S1_B					
H1_S1_B:H13_S1_B					
H2_S1_B:H2_S1_B	15.39	1.0222	4.4092	1.9643	9.8971
H2_S1_B:H3_S1_B	0.01	0.9996	0.9775	0.5326	1.7941
H2_S1_B:H4_S1_B	0.86	1.0055	1.311	0.739	2.326
H2_S1_B:H5_S1_B	0.29	0.9975	0.8237	0.4054	1.6738
H2_S1_B:H6_S1_B	3.48	1.008	2.2297	0.9393	5.2926
H2_S1_B:H7_S1_B	2.06	1.0058	1.8911	0.7814	4.5767
H2_S1_B:H8_S1_B					
H2_S1_B:H9_S1_B					
H2_S1_B:H10_S1_B					
H3_S1_B:H3_S1_B	14.34	0.9851	0.1606	0.0545	0.4734
H3_S1_B:H4_S1_B	11.15	0.9838	0.3047	0.1457	0.6371
H3_S1_B:H5_S1_B	12.6	0.9884	0.1093	0.0248	0.482
H3_S1_B:H6_S1_B	4.13	0.9933	0.3495	0.1211	1.0091
H3_S1_B:H7_S1_B					
H3_S1_B:H8_S1_B		0.9961	0.1542	0.018	1.3221
H3_S1_B:H9_S1_B		0.9992	0.7735	0.193	3.1001
H4_S1_B:H4_S1_B	1	1.0033	1.7088	0.5919	4.933
H4_S1_B:H5_S1_B	5.06	0.9924	0.3201	0.1124	0.9115
H4_S1_B:H6_S1_B		1.0063	7.7907	0.9957	60.9556
H4_S1_B:H7_S1_B		0.9999	0.9676	0.2592	3.6121
H5_S1_B:H5_S1_B					
H5_S1_B:H6_S1_B	1.04	0.9966	0.6001	0.2228	1.6166
H5_S1_B:H7_S1_B					

FIG. 47-2

Diplotype	Cases with	Cases w/o	Controls with	Controls w/o	p-value
H1_51_B:H1_51_B	84	1302	26	1047	<.0001
H1_51_B:H2_51_B	114	1272	46	1027	<.0001
H1_51_B:H3_51_B	43	1343	49	1024	0.05778
H1_51_B:H4_51_B	38	1348	25	1048	0.52197
H1_51_B:H5_51_B	22	1364	32	1041	0.019235
H1_51_B:H6_51_B	20	1366	18	1055	0.63904
H1_51_B:H7_51_B	14	1372	7	1066	0.340114
H1_51_B:H8_51_B	16	1370	5	1068	0.065593
H1_51_B:H9_51_B	11	1375	8	1065	
H1_51_B:H10_51_B	18	1368	4	1069	
H1_51_B:H11_51_B	15	1371	6	1067	
H1_51_B:H12_51_B	7	1379	5	1068	
H1_51_B:H13_51_B	10	1376	1	1072	
H2_51_B:H2_51_B	39	1347	7	1066	<.0001
H2_51_B:H3_51_B	24	1362	19	1054	0.920344
H2_51_B:H4_51_B	32	1354	19	1054	0.353739
H2_51_B:H5_51_B	16	1370	15	1058	0.590221
H2_51_B:H6_51_B	20	1366	7	1066	0.062115
H2_51_B:H7_51_B	17	1369	7	1066	0.15121
H2_51_B:H8_51_B	8	1378	5	1068	
H2_51_B:H9_51_B	9	1377	3	1070	
H2_51_B:H10_51_B	11	1375	1	1072	
H3_51_B:H3_51_B	4	1382	19	1054	0.000153
H3_51_B:H4_51_B	10	1376	25	1048	0.00084
H3_51_B:H5_51_B	2	1384	14	1059	0.000386
H3_51_B:H6_51_B	5	1381	11	1062	0.042129
H3_51_B:H7_51_B	0	1386	7	1066	
H3_51_B:H8_51_B	1	1385	5	1068	
H3_51_B:H9_51_B	4	1382	4	1069	
H4_51_B:H4_51_B	11	1375	5	1068	0.317311
H4_51_B:H5_51_B	5	1381	12	1061	0.024484
H4_51_B:H6_51_B	10	1376	1	1072	
H4_51_B:H7_51_B	5	1381	4	1069	
H5_51_B:H5_51_B	0	1386	5	1068	
H5_51_B:H6_51_B	7	1379	9	1064	0.307821
H5_51_B:H7_51_B	6	1380	4	1069	

FIG. 48-1

Diplotype	Chi Square (Pearson)	Risk Ratio	Odds Ratio	95% CI lower	95% CI upper
H1_S1_B:H1_S1_B	18.73	1.0387	2.598	1.661	4.0636
H1_S1_B:H2_S1_B	15.42	1.0429	2.0009	1.4071	2.8454
H1_S1_B:H3_S1_B	3.6	0.9849	0.6691	0.4407	1.0159
H1_S1_B:H4_S1_B	0.41	1.0042	1.1817	0.7088	1.9702
H1_S1_B:H5_S1_B	5.48	0.9858	0.5247	0.3031	0.9083
H1_S1_B:H6_S1_B	0.22	0.9976	0.8581	0.4517	1.6304
H1_S1_B:H7_S1_B	0.91	1.0036	1.5539	0.625	3.8637
H1_S1_B:H8_S1_B	3.39	1.007	2.4946	0.911	6.8313
H1_S1_B:H9_S1_B					
H1_S1_B:H10_S1_B					
H1_S1_B:H11_S1_B					
H1_S1_B:H12_S1_B					
H1_S1_B:H13_S1_B					
H2_S1_B:H2_S1_B	15.39	1.0222	4.4092	1.9643	9.8971
H2_S1_B:H3_S1_B	0.01	0.9996	0.9775	0.5326	1.7941
H2_S1_B:H4_S1_B	0.86	1.0055	1.311	0.739	2.326
H2_S1_B:H5_S1_B	0.29	0.9975	0.8237	0.4054	1.6738
H2_S1_B:H6_S1_B	3.48	1.008	2.2297	0.9393	5.2926
H2_S1_B:H7_S1_B	2.06	1.0058	1.8911	0.7814	4.5767
H2_S1_B:H8_S1_B					
H2_S1_B:H9_S1_B					
H2_S1_B:H10_S1_B					
H3_S1_B:H3_S1_B	14.34	0.9851	0.1606	0.0545	0.4734
H3_S1_B:H4_S1_B	11.15	0.9838	0.3047	0.1457	0.6371
H3_S1_B:H5_S1_B	12.6	0.9884	0.1093	0.0248	0.482
H3_S1_B:H6_S1_B	4.13	0.9933	0.3495	0.1211	1.0091
H3_S1_B:H7_S1_B					
H3_S1_B:H8_S1_B		0.9961	0.1542	0.018	1.3221
H3_S1_B:H9_S1_B		0.9992	0.7735	0.193	3.1001
H4_S1_B:H4_S1_B	1	1.0033	1.7088	0.5919	4.933
H4_S1_B:H5_S1_B	5.06	0.9924	0.3201	0.1124	0.9115
H4_S1_B:H6_S1_B		1.0063	7.7907	0.9957	60.9556
H4_S1_B:H7_S1_B		0.9999	0.9676	0.2592	3.6121
H5_S1_B:H5_S1_B					
H5_S1_B:H6_S1_B	1.04	0.9966	0.6001	0.2228	1.6166
H5_S1_B:H7_S1_B					

FIG. 48-2