# APPENDIX B

# SUPPLEMENTAL MATERIAL FOR
# CHAPTER 7.0.
# GENETIC INFLUENCE OF *SCARB1* VARIANTS ON LIPID TRAITS IN AFRICAN BLACKS

**Table B1. Characteristics and lipid profile of the entire African Black sample stratified by sex.**

| Variables | Males[a] | Females[a] | The Entire Sample[a] |
|---|---|---|---|
| **N** | 495 | 293 | 788 |
| **Age, years** | 42.35 ± 8.62 | 38.60 ± 7.43 | 40.95 ± 8.39 |
| **BMI, kg/m$^2$** | 22.03 ± 3.31 | 24.31 ± 4.71 | 22.87 ± 4.04 |
| **Total Cholesterol, mg/dL** | 166.65 ± 37.37 | 181.02 ± 38.69 | 172.01 ± 38.47 |
| **LDL-Cholesterol, mg/dL** | 104.52 ± 33.71 | 117.20 ± 34.14 | 109.25 ± 34.40 |
| **HDL-Cholesterol, mg/dL** | 46.03 ± 12.61 | 51.00 ± 12.71 | 47.88 ± 12.87 |
| **Triglycerides, mg/dL** | 78.72 ± 42.13 | 63.27 ± 31.88 | 72.96 ± 39.32 |
| **Apolipoprotein A-I, mg/dL** | 136.13 ± 29.37 | 138.54 ± 26.85 | 137.03 ± 28.46 |
| **Apolipoprotein B, mg/dL** | 65.35 ± 21.71 | 69.69 ± 22.74 | 66.98 ± 22.19 |

BMI, body mass index; HDL-Cholesterol, high-density lipoprotein cholesterol; LDL-Cholesterol, low-density lipoprotein cholesterol.

Values are presented as unadjusted means ± standard deviation (SD) unless otherwise mentioned.

[a] All data were unadjusted and included individuals with missing values or outliers (values beyond mean ± 3.5 SD).

**Table B2.** **Primer sequences and sizes of 14 polymerase chain reaction (PCR) fragments, and sizes of 13 *SCARB1* exons.**

| Fragment | Forward Primer Sequence | Reverse Primer Sequence | Size of PCR Fragment (bp) | Size of Exon (bp) |
|---|---|---|---|---|
| **Exon 1** | 5′-CCCATAGACGTTTTGGCTCA-3′ | 5′-AGCAACCTGTCCACACTCCT-3′ | 896 | 379 |
| **Exon 2** | 5′-TCCCTGCTGTGTTTTCTCTG-3′ | 5′-CTGGTGATTCGCACCTGTAA-3′ | 844 | 158 |
| **Exon 3** | 5′-TGGCTTGGAGAGATGAGAGA-3′ | 5′-TGGGAAACTCAGAACCCACT-3′ | 752 | 142 |
| **Exon 4** | 5′-CTGTCTTGTTGAGGGCTGAG-3′ | 5′-CAGGTTGCCAAGAGTCAAGA-3′ | 848 | 204 |
| **Exon 5** | 5′-CTCCTAGAAAGCTCCCAAGC-3′ | 5′-TTCACCTCCTGTGTTCAAGC-3′ | 791 | 96 |
| **Exon 6** | 5′-CTCACCTGCTCACCACACTT-3′ | 5′-TGATGTCTCAGCACCCAGAT-3′ | 770 | 116 |
| **Exon 7** | 5′-ACAGGTGTGAGCCATAATGC-3′ | 5′-GGTATCCAGGAGAGCACGAT-3′ | 989 | 167 |
| **Exon 8** | 5′-AGGGACACACTCCTGTGGAT-3′ | 5′-ACAGAACTTCACACGGGACA-3′ | 1,042 | 119 |
| **Exon 9** | 5′-TCAGGAGAGGAGATCCCAGT-3′ | 5′-CGTGTAGGAAACAGCTTGGA-3′ | 906 | 74 |
| **Exon 10** | 5′-TGAGGGTAAGAAATGGCAGA-3′ | 5′-ACAAGCTAGGCCAGAAGGAA-3′ | 862 | 52 |
| **Exon 11** | 5′-TCCTTCTGGCCTAGCTTGTT-3′ | 5′-GGTGCTGACTTGATGAATGG-3′ | 990 | 147 |
| **Exon 12** | 5′-ACATGCGGGTAAACTCAACA-3′ | 5′-AGCCTTGCTCCTGTCTTTCT-3′ | 1,033 | 129 |
| **Exon 13 (1)** | 5′-GCTCTGCCCCTCACTGTATT-3′ | 5′-TGTCAGTTTAGGCTGGAGGA-3′ | 978 | 959 |
| **Exon 13 (2)** | 5′-ATGGAGTGAGCACAAGATGC-3′ | 5′-TAAGGGGAAAAGGGCTAACA-3′ | 1,038 | |

RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics, University of Florida, http://snpper.chip.org/).

**Table B3.** **Characteristics of 83 *SCARB1* sequence variants identified in 95 African Blacks with extreme HDL-C levels.**

| | | | | | | | | | SEQ MAF | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | HWE *P* | %Call Rate | Total 95 AB Subjects | High HDL-C Group (48 ABs) | Low HDL-C Group (47 ABs) | Selection for GT (n = 78) | Success in GT (n = 68) | Note |
| p972 | rs181338950 | 125348548 | 5' flanking-promoter | | | C/T | 1 | 97.9 | 0.048 | 0.053 | 0.043 | Y | N | Failed GT assay design |
| p1048insC (1048_1049) | | 125348472 | Exon 1-5' UTR | | insC | [-/C] | 1 | 98.9 | 0.005 | 0 | **0.011** | Y | Y | |
| p1257 | rs4238001 | 125348263 | Exon 1 | Gly2Ser | | G/A | 0.1595 | 97.9 | 0.032 | 0.032 | 0.033 | Y | N | Failed GT run |
| p1265 | rs2070242 | 125348255 | Exon 1 | Ser4Ser | | C/T | 0.4979 | 94.7 | 0.167 | 0.109 | 0.227 | Y | Y | |
| p1316 | rs10396208 | 125348204 | Exon 1 | Cys21Cys | | C/T | 0.5545 | 94.7 | 0.061 | 0.033 | 0.091 | Y | Y | |
| p1419 | rs201717369 | 125348101 | Intron 1 | | | G/A | 1 | 98.9 | 0.021 | 0.021 | 0.022 | Y | Y | |
| p46964 | rs114061302 | 125302556 | Intron 1 | | | G/A | 1 | 100 | 0.032 | 0.031 | 0.032 | Y | Y | |
| p49537 | rs7305310 | 125299983 | Intron 2 | | | C/T | 0.8226 | 100 | 0.095 | 0.094 | 0.096 | Y | Y | |
| p49570delC | rs145376237 | 125299950 | Intron 2 | | delC | [C/-] | 0.5967 | 100 | 0.242 | 0.250 | 0.234 | Y | Y | |
| p49690 | rs4765615 | 125299830 | Intron 2 | | | G/A | 0.1327 | 100 | 0.447 | 0.365 | 0.532 | Y | Y | |
| p49759 | rs146272788 | 125299761 | Intron 2 | | | C/T | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y | |
| p49978 | rs5891 | 125299542 | Exon 3 | Val135Ile | | G/A | 1 | 100 | 0.011 | **0.021** | 0 | Y | Y | |
| p50024 | rs368880622 | 125299496 | Intron 3 | | | G/T | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y | |
| p50118 | rs58710319 | 125299402 | Intron 3 | | | C/T | 1 | 100 | 0.021 | 0.021 | 0.021 | Y | Y | |
| p50151 | rs2278986 | 125299369 | Intron 3 | | | T/C | 1 | 98.9 | 0.176 | 0.223 | 0.128 | Y | Y | |
| p50380 | rs141748317 | 125299140 | Intron 3 | | | A/G | 1 | 100 | 0.011 | 0.010 | 0.011 | Y | Y | |
| p50489 | rs61320152 | 125299031 | Intron 3 | | | G/T | 1 | 100 | 0.021 | 0.021 | 0.021 | Y | Y | |
| p50954 | | 125298566 | Intron 4 | | | T/C | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y | |
| p52919 | | 125296601 | Intron 4 | | | G/T | 1 | 100 | 0.011 | 0 | **0.021** | Y | Y | |
| p52956 | rs77740046 | 125296564 | Intron 4 | | | C/T | 1 | 100 | 0.047 | 0.052 | 0.043 | Y | Y | |
| p52995 | rs113910315 | 125296525 | Intron 4-splice site | | | T/G | 1 | 100 | 0.005 | 0 | **0.011** | Y | Y | |
| p53128 | rs372212527 | 125296392 | Intron 5 | | | C/T | 1 | 95.8 | 0.005 | **0.011** | 0 | Y | N | Failed GT run |
| p53159 | rs59809936 | 125296361 | Intron 5 | | | G/A | 1 | 95.8 | 0.049 | 0.054 | 0.044 | N | | Not selected, tagged with p52956[d] |
| p53359 | rs112371713 | 125296161 | Intron 5 | | | G/A | 1 | 100 | 0.121 | 0.156 | 0.085 | Y | Y | |
| p53372 | rs115604379 | 125296148 | Intron 5 | | | C/T | 1 | 100 | 0.016 | **0.031** | 0 | Y | Y | |
| p53481 | rs143611171 | 125296039 | Intron 5 | | | C/T | 0.7877 | 95.8 | 0.099 | 0.109 | 0.089 | Y | N | Failed GT run |
| p54445 | rs60910935 | 125295075 | Intron 5 | | | A/G | 1 | 100 | 0.032 | 0.021 | 0.043 | Y | Y | |
| p54475 | rs60227139 | 125295045 | Intron 5 | | | C/T | 1 | 100 | 0.032 | 0.021 | 0.043 | Y | Y | |
| p54492 | rs61762481 | 125295028 | Intron 5 | | | G/A | 0.5776 | 100 | 0.111 | 0.062 | 0.160 | Y | Y | |

| | | | | | | | | | SEQ MAF | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | HWE *P* | %Call Rate | Total 95 AB Subjects | High HDL-C Group (48 ABs) | Low HDL-C Group (47 ABs) | Selection for GT (n = 78) | Success in GT (n = 68) | Note |
| p54611 | | 125294909 | Intron 5 | | | T/C | 1 | 100 | 0.005 | 0 | **0.011** | Y | Y | |
| p54627 | | 125294893 | Intron 5 | | | G/C | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y | |
| p54856 | | 125294664 | Intron 6 | | | C/T | 1 | 100 | 0.005 | 0 | **0.011** | Y | Y | |
| p56845 | rs838902 | 125292675 | Intron 6 | | | A/G | 0.6252 | 98.9 | 0.41 | 0.385 | 0.435 | Y | Y | |
| p57004 | rs187562853 | 125292516 | Intron 6 | | | G/A | 1 | 100 | 0.021 | **0.042** | 0 | Y | Y | |
| p57107 | rs5892 | 125292413 | Exon 7 | Phe301Phe | | C/T | 1 | 100 | 0.026 | 0.031 | 0.021 | Y | Y | |
| p57508 | rs71458866 | 125292012 | Intron 7 | | | G/A | 0.646 | 98.9 | 0.106 | 0.073 | 0.141 | Y | Y | |
| p57592 | rs838903 | 125291928 | Intron 7 | | | G/A | 0.1308 | 98.9 | 0.356 | 0.323 | 0.391 | Y | Y | |
| p64772 | rs5888 | 125284748 | Exon 8 | Ala350Ala | | C/T | 1 | 100 | 0.116 | 0.125 | 0.106 | Y | Y | |
| p64923 | rs838915 | 125284597 | Intron 8 | | | C/A | 0.2751 | 100 | 0.137 | 0.156 | 0.117 | Y | Y | |
| p69699 | rs10396210 | 125279821 | Intron 8-splice site | | | G/A | 1 | 98.9 | 0.138 | 0.096 | 0.181 | Y | Y | |
| p69994 | rs188094006 | 125279526 | Intron 9 | | | C/T | 1 | 100 | 0.005 | **0.010** | 0 | Y | N | Failed GT run |
| p69995delC | rs5801571 | 125279525 | Intron 9 | | delC | [C/-] | 0.7558 | 100 | 0.253 | 0.250 | 0.255 | Y | Y | |
| p70088 | rs117585141 | 125279432 | Intron 9 | | | G/A | 1 | 100 | 0.158 | 0.125 | 0.191 | Y | N | Failed GT run |
| p70129 | rs1814575 | 125279391 | Intron 9 | | | C/T | 1 | 100 | 0.047 | 0.052 | 0.043 | Y | N | Failed GT run |
| p70148 | rs183061101 | 125279372 | Intron 9 | | | T/C | 1 | 100 | 0.011 | 0.010 | 0.011 | Y | N | Failed GT run |
| p70201 | | 125279319 | Intron 9 | | | T/C | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y | |
| p70292 | rs150388176 | 125279228 | Intron 9 | | | C/T | 1 | 98.9 | 0.016 | 0 | **0.033** | Y | N | Failed GT assay design |
| p77181 | rs146246031 | 125272339 | Intron 9 | | | T/C | 1 | 98.9 | 0.005 | **0.010** | 0 | Y | Y | |
| p77250 | rs201901986 | 125272270 | Intron 9 | | | G/T | 0.8226 | 100 | 0.095 | 0.052 | 0.138 | N | | Not selected, tagged with p77251[d] |
| p77251 | rs34339961 | 125272269 | Intron 9 | | | A/T | 0.8954 | 97.9 | 0.091 | 0.043 | 0.138 | Y | Y | |
| p77381 | rs138499966 | 125272139 | Intron 9 | | | T/C | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y | |
| p77620 | rs377124254 | 125271900 | Intron 10 | | | G/A | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y | |
| p77682 | rs150082885 | 125271838 | Intron 10 | | | A/G | 0.0317 | 100 | 0.016 | 0.010 | 0.021 | Y | Y | |
| p77704 | | 125271816 | Intron 10 | | | C/A | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y | |
| p77842 | rs2272310 | 125271678 | Intron 10 | | | G/A | 1 | 100 | 0.084 | 0.094 | 0.074 | Y | Y | |
| p78255 | rs184052375 | 125271265 | Intron 10 | | | A/G | 1 | 100 | 0.016 | 0.021 | 0.011 | Y | Y | |
| p78402 | rs838898 | 125271118 | Intron 10 | | | G/A | 1 | 100 | 0.063 | 0.073 | 0.053 | Y | Y | |
| p78430 | rs838897 | 125271090 | Intron 10 | | | C/G | 0.3016 | 94.7 | 0.356 | 0.344 | 0.367 | Y | Y | |
| p78747 | rs2293440 | 125270773 | Intron 11 | | | T/C | 0.8126 | 96.8 | 0.315 | 0.244 | 0.383 | Y | Y | |
| p78791 | rs75289200 | 125270729 | Intron 11 | | | T/C | 1 | 98.9 | 0.043 | 0.043 | 0.043 | Y | Y | |
| p81863 | rs185445624 | 125267657 | Intron 11 | | | G/A | 1 | 100 | 0.005 | 0 | **0.011** | Y | Y | |

|  |  |  |  |  |  |  |  |  | SEQ MAF |  |  |  |  |  |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | HWE *P* | %Call Rate | Total 95 AB Subjects | High HDL-C Group (48 ABs) | Low HDL-C Group (47 ABs) | Selection for GT (n = 78) | Success in GT (n = 68) | Note |
| p82019 | rs838890 | 125267501 | Intron 11 |  |  | C/T | 1 | 100 | 0.032 | 0.031 | 0.032 | Y | Y |  |
| p82264 | rs141545424 | 125267256 | Exon 12 | Gly501Gly |  | C/A | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y |  |
| p82340 | rs77483223 | 125267180 | Intron 12 |  |  | G/A | 1 | 100 | 0.005 | 0 | **0.011** | Y | Y |  |
| p82369 | rs75446635 | 125267151 | Intron 12 |  |  | G/A | 1 | 100 | 0.032 | 0.042 | 0.021 | Y | Y |  |
| p82434 | rs838889 | 125267086 | Intron 12 |  |  | T/C | 1 | 100 | 0.032 | 0.031 | 0.032 | Y | Y |  |
| p86245 | rs188375019 | 125263275 | Intron 12 |  |  | C/T | 1 | 98.9 | 0.032 | 0.031 | 0.033 | Y | Y |  |
| p86276 | rs747155 | 125263244 | Intron 12 |  |  | C/T | 0.8101 | 98.9 | 0.122 | 0.160 | 0.085 | Y | Y |  |
| p86316 | rs701104 | 125263204 | Intron 12 |  |  | G/T | 1 | 100 | 0.032 | 0.021 | 0.043 | Y | Y |  |
| p86481 | rs701103 | 125263039 | Exon 13-3' UTR | Gly499Arg (isoform 2) |  | G/A | 0.4173 | 97.9 | 0.237 | 0.298 | 0.174 | Y | Y |  |
| p86967 | rs187492239 | 125262553 | Exon 13-3' UTR |  |  | A/G | 1 | 97.9 | 0.032 | 0.032 | 0.033 | Y | Y |  |
| p87011 | rs58032386 | 125262509 | Exon 13-3' UTR |  |  | C/T | 0.3196 | 100 | 0.126 | 0.104 | 0.149 | Y | Y |  |
| p87195 | rs10396211 | 125262325 | Exon 13-3' UTR |  |  | G/C | 0.8101 | 98.9 | 0.122 | 0.094 | 0.152 | N |  | Not selected, tagged with p87011[d] |
| p87266 | rs150512235 | 125262254 | Exon 13-3' UTR[e] |  |  | T/C | 1 | 100 | 0.032 | 0.042 | 0.021 | Y | Y |  |
| p87416 | rs838884 | 125262104 | 3' flanking |  |  | C/T | 0.6922 | 97.9 | 0.301 | 0.323 | 0.278 | N |  | Not selected, tagged with p87723[d] |
| p87459 |  | 125262061 | 3' flanking |  |  | C/T | 1 | 98.9 | 0.005 | **0.010** | 0 | Y | N | Failed GT run |
| p87611 | rs190688220 | 125261909 | 3' flanking |  |  | C/T | 1 | 100 | 0.032 | 0.031 | 0.032 | Y | Y |  |
| p87681 | rs838883 | 125261839 | 3' flanking |  |  | G/A | 1 | 100 | 0.032 | 0.021 | 0.043 | Y | Y |  |
| p87694 |  | 125261826 | 3' flanking |  |  | C/T | 1 | 100 | 0.005 | **0.010** | 0 | Y | Y |  |
| p87707 | rs838882 | 125261813 | 3' flanking |  |  | G/A | 0.9969 | 98.9 | 0.303 | 0.333 | 0.272 | N |  | Not selected, tagged with p87723[d] |
| p87723 | rs838881 | 125261797 | 3' flanking |  |  | C/T | 0.6922 | 97.9 | 0.301 | 0.326 | 0.277 | Y | Y |  |
| p87749 | rs76465225 | 125261771 | 3' flanking |  |  | G/A | 0.3707 | 100 | 0.095 | 0.094 | 0.096 | Y | Y |  |
| p87927 | rs838880 | 125261593 | 3' flanking |  |  | G/A | 1 | 100 | 0.226 | 0.250 | 0.202 | Y | Y |  |

AB, African Black; del, deletion; GT, genotyping; HDL-C, high-density lipoprotein cholesterol; HWE; Hardy-Weinberg equilibrium; Indels, insertion and deletion variations; ins, insertion; MAF, minor allele frequency; SEQ, sequencing; SNP, single nucleotide polymorphism; UTR, untranslated region. N, no; Y, yes.

All alleles on reverse strand. Splice site is defined as ± 20 bp from the start or end of an exon.

The MAFs of sequence variants specific to one of the two extreme HDL-C groups are shown in **bold**.

[a, c] Based on the RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics).

[b] dbSNP build 139: GRCh37.p10; 10 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_AB).

[d] Linkage disequilibrium structures of a total of 83 sequence variants and 32 sequence variants with MAF ≥5% are shown in Figures B1 and B2, respectively.

[e] Close to the miRNA-145 seed binding site based on TargetScanHuman (version 6.2, http://www.targetscan.org/).

### Table B4. Characteristics of 10 *SCARB1* novel[a] variants.

| SNP Name[b] | Chr12 Position[c] | Location | RegulomeDB Score[d] | Major/Minor Alleles | MAF | MA Effect on Traits[e] |
|---|---|---|---|---|---|---|
| p1048insC (1048_1049) | 125348472 | Exon 1- 5' UTR | 2a | [-/C] | 0.0079 | |
| p50954 | 125298566 | Intron 4 | 5 | T/C | 0.0007 | |
| p52919 | 125296601 | Intron 4 | 5 | G/T | 0.0013 | ↓ HDL-C, ↓ ApoA-I |
| p54611 | 125294909 | Intron 5 | 4 | T/C | 0.0007 | ↓ HDL-C, ↓ ApoA-I |
| p54627 | 125294893 | Intron 5 | 4 | G/C | 0.0020 | |
| p54856 | 125294664 | Intron 6 | 4 | C/T | 0.0007 | ↓ HDL-C, ↓ ApoA-I |
| p70201[f] | 125279319 | Intron 9 | 6 | T/C | 0.0010 | |
| p77704 | 125271816 | Intron 10 | 5 | C/A | 0.0040 | |
| p87459 | 125262061 | 3' flanking | 4 | C/T | 0.005[g] | |
| p87694 | 125261826 | 3' flanking | 5 | C/T | 0.0020 | |

ApoA-I, apolipoprotein A-I; HDL-C, high-density lipoprotein cholesterol; ins, insertion; MA, minor allele; MAF, minor allele frequency; SNP; single nucleotide polymorphism; UTR, untranslated region. ↓, decreased.

All alleles on reverse strand.

[a] dbSNP build 139: GRCh37.p10; 10 novel variants were submitted to dbSNP: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_AB).

[b, c] Based on the RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics).

[d] The RegulomeDB (version 1.0) scoring scheme represented as following: **score 1a**, expression quantitative trait loci (eQTL) + transcription factor (TF) binding + matched TF motif + matched DNase Footprint + DNase peak; **score 1b**, eQTL + TF binding + any motif + DNase Footprint + DNase peak; **score 1c**, eQTL + TF binding + matched TF motif + DNase peak; **score 1d**, eQTL + TF binding + any motif + DNase peak; **score 1e**, eQTL + TF binding + matched TF motif; **score 1f**, eQTL + TF binding / DNase peak, **score 2a**, TF binding + matched TF motif + matched DNase Footprint + DNase peak; **score 2b**, TF binding + any motif + DNase Footprint + DNase peak; **score 2c**, TF binding + matched TF motif + DNase peak; **score 3a**, TF binding + any motif + DNase peak; **score 3b**, TF binding + matched TF motif; **score 4**,TF binding + DNase peak; **score 5**, TF binding or DNase peak; **score 6**, others; **score 7**, no data, or can be seen at http://regulome.stanford.edu/help. See detailed functional assignments in Table B13.

[e] Allele effects are based on single-site association results, see details in Tables B9 and B10.

[f] p70201-chr12_125279319 was excluded from gene-based, single-site, and haplotype analyses, but included in rare variant analysis; see details in Table B6 and Figure 7.1.

[g] Derived from sequencing data due to genotyping failure.

**Table B5. List of 77 *SCARB1* common HapMap-YRI tagSNPs (HapMap release #27, genome build 36, dbSNP version 126) identified by Haploview Tagger analysis at $r^2$ ≥0.80, which captured a total of 108 common HapMap-YRI tagSNPs (MAF ≥5%) with a mean $r^2$ of 0.98 (see linkage disequilibrium structure of 108 *SCARB1* common HapMap-YRI tagSNPs in Figure B3).**

| Bin # | TagSNP | Alleles Captured |
|---|---|---|
| 1 | rs9919713 | rs838867,rs838865,rs1672879,rs701107 |
| 2 | rs838893 | rs838892,rs838894,rs838891 |
| 3 | rs4765178 | rs4765616,rs4765618,rs7135223 |
| 4 | **rs838881** | rs838882,rs838879,rs838884 |
| 5 | rs10846751 | rs4765623,rs10846748,rs4765621 |
| 6 | rs2343394 | rs2278986, rs745529 |
| 7 | rs838912[a] | rs5888[a],rs10773105 |
| 8 | rs12819677 | rs989892,rs3782287 |
| 9 | rs7137797 | rs7306660 |
| 10 | rs4765614 | rs4765613 |
| 11 | rs10773111 | rs7954697 |
| 12 | rs12370382 | rs12582221 |
| 13 | rs11057844 | rs7954519 |
| 14 | rs838905 | rs11057818 |
| 15 | rs12229555 | rs10846753 |
| 16 | rs4765622 | rs4765624 |
| 17 | rs961170 | rs961169 |
| 18 | rs3924313 | |
| 19 | rs838866 | |
| 20 | rs10846744 | |
| 21 | rs10744181 | |
| 22 | rs4765181 | |
| 23 | rs7135117 | |
| 24 | rs7138304 | |
| 25 | rs838887 | |
| 26 | rs12581963 | |
| 27 | rs11057853[a] | |
| 28 | rs11608336 | |
| 29 | rs10846749 | |
| 30 | rs838896[a] | |
| 31 | rs6488944 | |
| 32 | rs10773112 | |
| 33 | **rs4765615**[a] | |
| 34 | rs11057869 | |
| 35 | rs7954022 | |
| 36 | rs11057852 | |
| 37 | rs6488943 | |
| 38 | rs11057820 | |
| 39 | rs10744182 | |

|  |  |  |
|---|---|---|
| 40 | rs838861 |  |
| 41 | rs10846745 |  |
| 42 | rs838911 |  |
| 43 | rs4765180[d] |  |
| 44 | **rs10396210** |  |
| 45 | rs838862 |  |
| 46 | rs7967521 |  |
| 47 | **rs2272310** |  |
| 48 | rs865716 |  |
| 49 | rs12297372 |  |
| 50 | rs7134858[a] |  |
| 51 | rs7139401 |  |
| 52 | rs1726374 |  |
| 53 | rs12580803 |  |
| 54 | rs701106[a] |  |
| 55 | rs838909 |  |
| 56 | rs11057864 |  |
| 57 | **rs5892** |  |
| 58 | rs10846739 |  |
| 59 | **rs838880** |  |
| 60 | rs866793[c] |  |
| 61 | **rs2293440** |  |
| 62 | rs10846738 |  |
| 63 | rs838910 |  |
| 64 | rs12425134 |  |
| 65 | rs838900 |  |
| 66 | rs1902569[a] |  |
| 67 | rs10773107 |  |
| 68 | rs10396211[b] |  |
| 69 | rs11615630 |  |
| 70 | rs7301120 |  |
| 71 | rs11057838 |  |
| 72 | rs7967406 |  |
| 73 | rs7138386 |  |
| 74 | rs12831105 |  |
| 75 | rs11057830 |  |
| 76 | rs11057841 |  |
| 77 | rs11057851[a] |  |

MAF, minor allele frequency; SNP, single nucleotide polymorphism; YRI, Yoruba people of Ibadan, Nigeria.

Of total 77 common HapMap-YRI tagSNPs, 69 were selected for genotyping in addition to seven tagSNPs that were already selected (Table B3) based on our sequencing data (shown in **bold**), and one was tagged by another already selected sequence variant in our data (see below under “b”).

Of theses 69 common HapMap-YRI tagSNPs selected for genotyping, 68 were successfully genotyped, and 67 were advance into downstream analyses (Table B6); see below under “c” the variant that was excluded from downstream analyses as it failed quality control checks and see below under “d” the variant that failed genotyping.

[a] SNPs with nominally significant evidence of single-site association with either high-density lipoprotein cholesterol or apolipoprotein A-I (*P* <0.05; Tables B9 and B10) observed in the current study.

[b] rs10396211 was not selected for genotyping. rs10396211 was tagged by one SNP identified by sequencing, rs58032386 ($r^2$ = 0.95), in our sample (see enlarged view B in Figure B2).

[c] rs866793 was deviated from Hardy-Weinberg equilibrium and excluded from downstream analyses.

[d] rs4765180 failed genotyping and excluded from downstream analyses.

## Table B6. Characteristics of 138 *SCARB1* genotyped variants in the entire sample of 788 African Blacks.

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino acid Change | Major/Minor Alleles | HWE *P* | %Call Rate | MAF | Source of Variants | Success in GT (n = 138) | Proceed to Analyses (n = 137) | Note |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p1048insC (1048_1049) | | 125348472 | Exon 1-5' UTR | | [-/C] | 1 | 96.1 | 0.0079 | SEQ | Y | Y | |
| p1265 | rs2070242 | 125348255 | Exon 1 | Ser4Ser | C/T | 0.8725 | 93.4 | 0.1284 | SEQ | Y | Y | |
| p1316 | rs10396208 | 125348204 | Exon 1 | Cys21Cys | C/T | 0.1503 | 93.4 | 0.0476 | SEQ | Y | Y | |
| p1419 | rs201717369 | 125348101 | Intron 1 | | G/A | 0.1976 | 94.4 | 0.0121 | SEQ | Y | Y | |
| p4072 | rs7139401 | 125345448 | Intron 1 | | T/C | 0.331 | 95.1 | 0.4386 | HapMap | Y | Y | |
| p5055 | rs11057869 | 125344465 | Intron 1 | | G/A | 0.0974 | 97.7 | 0.0740 | HapMap | Y | Y | |
| p6600 | rs12831105 | 125342920 | Intron 1 | | C/T | 0.979 | 95.1 | 0.1188 | HapMap | Y | Y | |
| p7650 | rs11615630 | 125341870 | Intron 1 | | G/A | 0.4345 | 99 | 0.0436 | HapMap | Y | Y | |
| p10292 | rs4765181 | 125339228 | Intron 1 | | G/T | 0.6396 | 98.4 | 0.2490 | HapMap | Y | Y | |
| p10991 | rs10773112 | 125338529 | Intron 1 | | G/A | 0.8029 | 95.7 | 0.3534 | HapMap | Y | Y | |
| p13570 | rs11057864 | 125335950 | Intron 1 | | G/T | 0.193 | 97.8 | 0.1180 | HapMap | Y | Y | |
| p16565 | rs10773111 | 125332955 | Intron 1 | | G/A | 0.3383 | 98.1 | 0.1928 | HapMap | Y | Y | |
| p20207 | rs11057853 | 125329313 | Intron 1 | | G/A | 0.7053 | 97.2 | 0.4484 | HapMap | Y | Y | |
| p20694 | rs11057852 | 125328826 | Intron 1 | | G/A | 0.3519 | 94.3 | 0.1050 | HapMap | Y | Y | |
| p20741 | rs11057851 | 125328779 | Intron 1 | | C/T | 0.4728 | 96.8 | 0.3237 | HapMap | Y | Y | |
| p21145 | rs3924313 | 125328375 | Intron 1 | | C/T | 1 | 98.1 | 0.1772 | HapMap | Y | Y | |
| p22116 | rs12370382 | 125327404 | Intron 1 | | G/A | 1 | 96.4 | 0.0645 | HapMap | Y | Y | |
| p22168 | rs7137797 | 125327352 | Intron 1 | | T/C | 0.6804 | 94.9 | 0.3977 | HapMap | Y | Y | |
| p22331 | rs6488944 | 125327189 | Intron 1 | | T/G | 0.2943 | 94.3 | 0.1474 | HapMap | Y | Y | |
| p22675 | rs12425134 | 125326845 | Intron 1 | | G/T | 1 | 96.4 | 0.0526 | HapMap | Y | Y | |
| p28137 | rs12229555 | 125321383 | Intron 1 | | A/G | 0.0888 | 95.9 | 0.3896 | HapMap | Y | Y | |
| p28692 | rs4765622 | 125320828 | Intron 1 | | C/T | 0.2025 | 97 | 0.2565 | HapMap | Y | Y | |
| p28957 | rs11057844 | 125320563 | Intron 1 | | G/A | 0.9612 | 96.4 | 0.2362 | HapMap | Y | Y | |
| p29749 | rs10846751 | 125319771 | Intron 1 | | C/T | 0.6254 | 93.7 | 0.4492 | HapMap | Y | Y | |
| p31072 | rs10846749 | 125318448 | Intron 1 | | C/G | 0.9027 | 96.6 | 0.4461 | HapMap | Y | Y | |
| p31938 | rs10744182 | 125317582 | Intron 1 | | G/A | 0.9847 | 96.7 | 0.1837 | HapMap | Y | Y | |
| p32129 | rs10773107 | 125317391 | Intron 1 | | G/T | 0.7277 | 96.8 | 0.1009 | HapMap | Y | Y | |
| p32273 | rs12580803 | 125317247 | Intron 1 | | T/C | 2.00E-04 | 99 | 0.1006 | HapMap | Y | Y | |
| p32290 | rs10744181 | 125317230 | Intron 1 | | T/C | 0.0356 | 92.3 | 0.1238 | HapMap | Y | Y | |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino acid Change | Major/Minor Alleles | HWE *P* | %Call Rate | MAF | Source of Variants | Success in GT (n = 138) | Proceed to Analyses (n = 137) | Note |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p32395 | rs12581963 | 125317125 | Intron 1 | | C/T | 0.5734 | 98.5 | 0.1314 | HapMap | Y | Y | |
| p32750 | rs7967521 | 125316770 | Intron 1 | | A/G | 0.2715 | 91.9 | 0.3425 | HapMap | Y | Y | |
| p32777 | rs11057841 | 125316743 | Intron 1 | | G/A | 0.3613 | 94.5 | 0.2805 | HapMap | Y | Y | |
| p32860 | rs7967406 | 125316660 | Intron 1 | | A/C | 0.6852 | 98.6 | 0.0991 | HapMap | Y | Y | |
| p33531 | rs11057838 | 125315989 | Intron 1 | | C/A | 0.7642 | 95.8 | 0.2278 | HapMap | Y | Y | |
| p36094 | rs11608336 | 125313426 | Intron 1 | | G/A | 0.7249 | 95.4 | 0.1543 | HapMap | Y | Y | |
| p36461 | rs4765178 | 125313059 | Intron 1 | | C/T | 0.8082 | 93.4 | 0.1671 | HapMap | Y | Y | |
| p36908 | rs10846745 | 125312612 | Intron 1 | | C/G | 0.6639 | 97.2 | 0.3257 | HapMap | Y | Y | |
| p37095 | rs10846744 | 125312425 | Intron 1 | | C/G | 0.0194 | 98.2 | 0.3056 | HapMap | Y | Y | |
| p41632 | rs6488943 | 125307888 | Intron 1 | | A/C | 0.0626 | 91.5 | 0.2954 | HapMap | Y | Y | |
| p42467 | rs11057830 | 125307053 | Intron 1 | | C/T | 0.3753 | 97.1 | 0.1523 | HapMap | Y | Y | |
| p45516 | rs1902569 | 125304004 | Intron 1 | | G/A | 1 | 94.9 | 0.1544 | HapMap | Y | Y | |
| p45627 | rs12297372 | 125303893 | Intron 1 | | A/G | 0.9301 | 95.1 | 0.0487 | HapMap | Y | Y | |
| p46964 | rs114061302 | 125302556 | Intron 1 | | G/A | 1 | 98.1 | 0.0388 | SEQ | Y | Y | |
| p48969 | rs2343394 | 125300551 | Intron 2 | | C/T | 0.401 | 98.6 | 0.1898 | HapMap | Y | Y | |
| p49537 | rs7305310 | 125299983 | Intron 2 | | C/T | 0.0033 | 95.2 | 0.1007 | SEQ | Y | Y | |
| p49570delC | rs145376237 | 125299950 | Intron 2 | | [C/-] | 0.8066 | 95.1 | 0.2276 | SEQ | Y | Y | |
| p49690 | rs4765615 | 125299830 | Intron 2 | | G/A | 0.0084 | 93.9 | 0.4426 | SEQ | Y | Y | |
| p49759 | rs146272788 | 125299761 | Intron 2 | | C/T | 1 | 95.2 | 0.0020 | SEQ | Y | Y | |
| p49978 | rs5891 | 125299542 | Exon 3 | Val135Ile | G/A | 1 | 99.1 | 0.0058 | SEQ | Y | Y | |
| p50024 | rs368880622 | 125299496 | Intron 3 | | G/T | 1 | 96.7 | 0.0026 | SEQ | Y | Y | |
| p50118 | rs58710319 | 125299402 | Intron 3 | | C/T | 1 | 97.6 | 0.0208 | SEQ | Y | Y | |
| p50151 | rs2278986 | 125299369 | Intron 3 | | T/C | 0.3798 | 97.8 | 0.1933 | SEQ | Y | Y | |
| p50380 | rs141748317 | 125299140 | Intron 3 | | A/G | 1 | 96.4 | 0.0112 | SEQ | Y | Y | |
| p50489 | rs61320152 | 125299031 | Intron 3 | | G/T | 1 | 96.3 | 0.0257 | SEQ | Y | Y | |
| p50954 | | 125298566 | Intron 4 | | T/C | 1 | 96.2 | 0.0007 | SEQ | Y | Y | |
| p51888 | rs7138304 | 125297632 | Intron 4 | | C/T | 0.0164 | 95.8 | 0.1079 | HapMap | Y | Y | |
| p52096 | rs10846739 | 125297424 | Intron 4 | | A/G | 0.0939 | 95.2 | 0.4693 | HapMap | Y | Y | |
| p52556 | rs11057820 | 125296964 | Intron 4 | | G/A | 0.1399 | 98.4 | 0.1000 | HapMap | Y | Y | |
| p52610 | rs10846738 | 125296910 | Intron 4 | | C/T | 0.9717 | 95.9 | 0.1349 | HapMap | Y | Y | |
| p52919 | | 125296601 | Intron 4 | | G/T | 1 | 96.2 | 0.0013 | SEQ | Y | Y | |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino acid Change | Major/Minor Alleles | HWE *P* | %Call Rate | MAF | Source of Variants | Success in GT (n = 138) | Proceed to Analyses (n = 137) | Note |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p52956 | rs77740046 | 125296564 | Intron 4 |  | C/T | 0.7667 | 95.3 | 0.0546 | SEQ | Y | Y |  |
| p52995 | rs113910315 | 125296525 | Intron 4-splice site |  | T/G | 1 | 97.1 | 0.0020 | SEQ | Y | Y |  |
| p53359 | rs112371713 | 125296161 | Intron 5 |  | G/A | 0.7881 | 93.9 | 0.1243 | SEQ | Y | Y |  |
| p53372 | rs115604379 | 125296148 | Intron 5 |  | C/T | 1 | 96.6 | 0.0066 | SEQ | Y | Y |  |
| p53790 | rs4765614 | 125295730 | Intron 5 |  | G/A | 0.2998 | 95.7 | 0.2653 | HapMap | Y | Y |  |
| p54445 | rs60910935 | 125295075 | Intron 5 |  | A/G | 0.7209 | 92.6 | 0.0418 | SEQ | Y | Y |  |
| p54475 | rs60227139 | 125295045 | Intron 5 |  | C/T | 0.8434 | 95.8 | 0.0437 | SEQ | Y | Y |  |
| p54492 | rs61762481 | 125295028 | Intron 5 |  | G/A | 1 | 99.1 | 0.1005 | SEQ | Y | Y |  |
| p54611 |  | 125294909 | Intron 5 |  | T/C | 1 | 97.1 | 0.0007 | SEQ | Y | Y |  |
| p54627 |  | 125294893 | Intron 5 |  | G/C | 1 | 97 | 0.0020 | SEQ | Y | Y |  |
| p54856 |  | 125294664 | Intron 6 |  | C/T | 1 | 97.1 | 0.0007 | SEQ | Y | Y |  |
| p55923 | rs838900 | 125293597 | Intron 6 |  | G/A | 0.6724 | 96.4 | 0.3921 | HapMap | Y | Y |  |
| p55963 | rs7134858 | 125293557 | Intron 6 |  | C/T | 0.189 | 97.6 | 0.1560 | HapMap | Y | Y |  |
| p56845 | rs838902 | 125292675 | Intron 6 |  | A/G | 0.2799 | 97.2 | 0.4249 | SEQ | Y | Y |  |
| p57004 | rs187562853 | 125292516 | Intron 6 |  | G/A | 1 | 97 | 0.0098 | SEQ | Y | Y |  |
| p57107 | rs5892 | 125292413 | Exon 7 | Phe301Phe | C/T | 0.4893 | 97 | 0.0589 | SEQ | Y | Y |  |
| p57508 | rs71458866 | 125292012 | Intron 7 |  | G/A | 0.3467 | 98.9 | 0.1130 | SEQ | Y | Y |  |
| p57592 | rs838903 | 125291928 | Intron 7 |  | G/A | 0.3215 | 97 | 0.3763 | SEQ | Y | Y |  |
| p58514 | rs838905 | 125291006 | Intron 7 |  | T/C | 0.2442 | 97.3 | 0.4329 | HapMap | Y | Y |  |
| p58664 | rs865716 | 125290856 | Intron 7 |  | A/T | 0.1006 | 94.7 | 0.2708 | HapMap | Y | Y |  |
| p60255 | rs3782287 | 125289265 | Intron 7 |  | C/T | 0.4661 | 97.7 | 0.2831 | Literature | Y | Y |  |
| p61872 | rs838909 | 125287648 | Intron 7 |  | C/T | 0.9762 | 95.8 | 0.2199 | HapMap | Y | Y |  |
| p62140 | rs838910 | 125287380 | Intron 7 |  | G/T | 0.7286 | 95.2 | 0.3047 | HapMap | Y | Y |  |
| p62409 | rs838911 | 125287111 | Intron 7 |  | C/T | 0.5513 | 94.9 | 0.4211 | HapMap | Y | Y |  |
| p62615 | rs7138386 | 125286905 | Intron 7 |  | T/C | 0.4533 | 93.8 | 0.1137 | HapMap | Y | Y |  |
| p63483 | rs838912 | 125286037 | Intron 7 |  | G/A | 1 | 97.3 | 0.0867 | HapMap | Y | Y |  |
| p64772 | rs5888 | 125284748 | Exon 8 | Ala350Ala | C/T | 0.823 | 97.7 | 0.0961 | SEQ | Y | Y |  |
| p64923 | rs838915 | 125284597 | Intron 8 |  | C/A | 0.3967 | 96.8 | 0.1435 | SEQ | Y | Y |  |
| p65999 | rs12819677 | 125283521 | Intron 8 |  | G/A | 0.2879 | 95.4 | 0.2813 | HapMap | Y | Y |  |
| p67439 | rs961170 | 125282081 | Intron 8 |  | G/A | 0.0532 | 95.2 | 0.0893 | HapMap | Y | Y |  |
| p67700 | rs1726374 | 125281820 | Intron 8 |  | G/A | 0.2011 | 98.5 | 0.1933 | HapMap | Y | Y |  |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino acid Change | Major/Minor Alleles | HWE *P* | %Call Rate | MAF | Source of Variants | Success in GT (n = 138) | Proceed to Analyses (n = 137) | Note |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p69013 | rs7135117 | 125280507 | Intron 8 | | A/G | 0.0258 | 94.9 | 0.2901 | HapMap | Y | Y | |
| p69699 | rs10396210 | 125279821 | Intron 8-splice site | | G/A | 1 | 94.9 | 0.1511 | SEQ | Y | Y | |
| p69995delC | rs5801571 | 125279525 | Intron 9 | | [C/-] | 0.2836 | 96.1 | 0.2761 | SEQ | Y | Y | |
| p70201 | | 125279319 | Intron 9 | | T/C | 1 | 97.1 | 0.0010 | SEQ | Y | Y-only in SKAT-O | Found in 1 subject with an outlier HDL-C value (above mean + 3.5 SD), then excluded from other analyses, except for SKAT-O test (see Figure 7.1). |
| p71867 | rs7954022 | 125277653 | Intron 9 | | C/T | 0.8535 | 95.9 | 0.1323 | HapMap | Y | Y | |
| p72197 | rs838861 | 125277323 | Intron 9 | | A/G | 0.0045 | 95.4 | 0.3777 | HapMap | Y | Y | |
| p72777 | rs838862 | 125276743 | Intron 9 | | C/T | 0.7348 | 95.2 | 0.0887 | HapMap | Y | Y | |
| p75766 | rs838866 | 125273754 | Intron 9 | | T/C | 0.4407 | 96.6 | 0.2116 | HapMap | Y | Y | |
| p75778 | rs7301120 | 125273742 | Intron 9 | | C/T | 1 | 93.9 | 0.1135 | HapMap | Y | Y | |
| p76204 | rs866793 | 125273316 | Intron 9 | | A/G | 1.66E-18 | 92.9 | 0.0520 | HapMap | Y | N | Excluded due to a deviation from HWE |
| p76757 | rs9919713 | 125272763 | Intron 9 | | A/T | 0.252 | 96.7 | 0.4390 | HapMap | Y | Y | |
| p77181 | rs146246031 | 125272339 | Intron 9 | | T/C | 1 | 96.2 | 0.0053 | SEQ | Y | Y | |
| p77251 | rs34339961 | 125272269 | Intron 9 | | A/T | 0.9341 | 95.4 | 0.1177 | SEQ | Y | Y | |
| p77381 | rs138499966 | 125272139 | Intron 9 | | T/C | 1 | 97 | 0.0046 | SEQ | Y | Y | |
| p77620 | rs377124254 | 125271900 | Intron 10 | | G/A | 1 | 96.2 | 0.0007 | SEQ | Y | Y | |
| p77682 | rs150082885 | 125271838 | Intron 10 | | A/G | 0.1549 | 95.4 | 0.0106 | SEQ | Y | Y | |
| p77704 | | 125271816 | Intron 10 | | C/A | 1 | 95.6 | 0.0040 | SEQ | Y | Y | |
| p77842 | rs2272310 | 125271678 | Intron 10 | | G/A | 1 | 98.2 | 0.0807 | SEQ | Y | Y | |
| p78255 | rs184052375 | 125271265 | Intron 10 | | A/G | 1 | 97.1 | 0.0072 | SEQ | Y | Y | |
| p78402 | rs838898 | 125271118 | Intron 10 | | G/A | 0.1137 | 89.7 | 0.0714 | SEQ | Y | Y | |
| p78430 | rs838897 | 125271090 | Intron 10 | | C/G | 0.0119 | 95.4 | 0.3830 | SEQ | Y | Y | |
| p78747 | rs2293440 | 125270773 | Intron 11 | | T/C | 0.5489 | 95.1 | 0.4112 | SEQ | Y | Y | |
| p78791 | rs75289200 | 125270729 | Intron 11 | | T/C | 0.9169 | 94.8 | 0.0321 | SEQ | Y | Y | |
| p79721 | rs838896 | 125269799 | Intron 11 | | G/C | 0.9259 | 97.7 | 0.3104 | HapMap | Y | Y | |
| p79828 | rs838895 | 125269692 | Intron 11 | | C/G | 0.8944 | 96.4 | 0.3171 | Literature | Y | Y | |
| p80045 | rs838893 | 125269475 | Intron 11 | | G/A | 0.9108 | 97.6 | 0.3244 | HapMap | Y | Y | |
| p81863 | rs185445624 | 125267657 | Intron 11 | | G/A | 1 | 97 | 0.0020 | SEQ | Y | Y | |
| p82019 | rs838890 | 125267501 | Intron 11 | | C/T | 0.343 | 95.1 | 0.0320 | SEQ | Y | Y | |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino acid Change | Major/Minor Alleles | HWE *P* | %Call Rate | MAF | Source of Variants | Success in GT (n = 138) | Proceed to Analyses (n = 137) | Note |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p82264 | rs141545424 | 125267256 | Exon 12 | Gly501Gly | C/A | 1 | 96.7 | 0.0007 | SEQ | Y | Y | |
| p82340 | rs77483223 | 125267180 | Intron 12 | | G/A | 1 | 95.9 | 0.0231 | SEQ | Y | Y | |
| p82369 | rs75446635 | 125267151 | Intron 12 | | G/A | 1 | 97 | 0.0059 | SEQ | Y | Y | |
| p82434 | rs838889 | 125267086 | Intron 12 | | T/C | 0.3344 | 96.6 | 0.0315 | SEQ | Y | Y | |
| p83547 | rs838887 | 125265973 | Intron 12 | | C/G | 0.5641 | 96.1 | 0.4564 | HapMap | Y | Y | |
| p83884 | rs701106 | 125265636 | Intron 12 | | C/T | 0.9346 | 98 | 0.2597 | HapMap | Y | Y | |
| p86245 | rs188375019 | 125263275 | Intron 12 | | C/T | 0.8126 | 96.7 | 0.0341 | SEQ | Y | Y | |
| p86276 | rs747155 | 125263244 | Intron 12 | | C/T | 0.8803 | 97.2 | 0.1495 | SEQ | Y | Y | |
| p86316 | rs701104 | 125263204 | Intron 12 | | G/T | 1 | 93.8 | 0.0487 | SEQ | Y | Y | |
| p86481 | rs701103 | 125263039 | Exon 13-3' UTR | Gly499Arg (isoform2) | G/A | 0.3376 | 96.6 | 0.2451 | SEQ | Y | Y | |
| p86967 | rs187492239 | 125262553 | Exon 13-3' UTR | | A/G | 0.7536 | 96.4 | 0.0355 | SEQ | Y | Y | |
| p87011 | rs58032386 | 125262509 | Exon 13-3' UTR | | C/T | 0.8106 | 97.6 | 0.1417 | SEQ | Y | Y | |
| p87266 | rs150512235 | 125262254 | Exon 13-3' UTR[d] | | T/C | 1 | 99.4 | 0.0057 | SEQ | Y | Y | |
| p87611 | rs190688220 | 125261909 | 3' flanking | | C/T | 0.9286 | 96.3 | 0.0316 | SEQ | Y | Y | |
| p87681 | rs838883 | 125261839 | 3' flanking | | G/A | 1 | 93.9 | 0.0459 | SEQ | Y | Y | |
| p87694 | | 125261826 | 3' flanking | | C/T | 1 | 95.4 | 0.0020 | SEQ | Y | Y | |
| p87723 | rs838881 | 125261797 | 3' flanking | | C/T | 0.0678 | 97.1 | 0.3183 | SEQ | Y | Y | |
| p87749 | rs76465225 | 125261771 | 3' flanking | | G/A | 0.5802 | 97 | 0.0844 | SEQ | Y | Y | |
| p87927 | rs838880 | 125261593 | 3' flanking | | G/A | 0.5951 | 96.4 | 0.2414 | SEQ | Y | Y | |

del, deletion; GT, genotyping; HDL-C, high-density lipoprotein cholesterol; HWE, Hardy-Weinberg equilibrium; ins, insertion; MAF, minor allele frequency; Literature, variants selected from the literature; SD, standard deviation; SEQ, sequencing; SKAT-O, an optimal sequence kernel association test; SNP, single nucleotide polymorphism; UTR, untranslated region. N, no; Y, yes.

All alleles on reverse strand. Splice site is defined as ± 20 bp from the start or end of an exon.

A total of 137 genotyped variants (68 variants identified by sequencing, 67 common HapMap-YRI tagSNPs (YRI, Yoruba people of Ibadan, Nigeria), and 2 additional relevant variants from the literature) that passed genotyping quality controls were included in downstream analyses.

A linkage disequilibrium structure of 137 genotyped variants that passed genotyping quality controls and were included in downstream analyses is shown in Figure B4.

Regional and MAF distributions of 137 genotyped variants is shown in Figure B5.

A list of 87 genotyped common tagSNPs (MAF ≥5%) is shown in Table B7.

[a, c] Based on the RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics).

[b] dbSNP build 139: GRCh37.p10; 10 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_AB).

[d] Close to the miRNA-145 seed binding site based on TargetScanHuman (version 6.2, http://www.targetscan.org/).

**Table B7. List of 87 *SCARB1* genotyped common tagSNPs identified by Haploview Tagger analysis at $r^2$ ≥0.90, which captured a total of 94 common tagSNPs (MAF ≥5%) that were genotyped in the entire sample of 788 African Blacks with a mean $r^2$ of 0.996.**

| Bin # | TagSNP | Alleles Captured |
|---|---|---|
| 1 | p1265-rs2070242 (p.Ser4Ser) | |
| 2 | p4072-rs7139401 | |
| 3 | p5055-rs11057869 | |
| 4 | p6600-rs12831105 | |
| 5 | p10292-rs4765181 | |
| 6 | p10991-rs10773112 | |
| 7 | p13570-rs11057864 | |
| 8 | p16565-rs10773111 | |
| 9 | **p20207-rs11057853** | |
| 10 | p20694-rs11057852 | |
| 11 | **p20741-rs11057851** | |
| 12 | p21145-rs3924313 | |
| 13 | p22116-rs12370382 | |
| 14 | p22168-rs7137797 | |
| 15 | p22331-rs6488944 | |
| 16 | p22675-rs12425134 | |
| 17 | p28137-rs12229555 | |
| 18 | p28692-rs4765622 | |
| 19 | p28957-rs11057844 | |
| 20 | p29749-rs10846751 | |
| 21 | p31072-rs10846749 | |
| 22 | p31938-rs10744182 | |
| 23 | p32129-rs10773107 | |
| 24 | p32273-rs12580803 | |
| 25 | p32290-rs10744181 | |
| 26 | p32395-rs12581963 | |
| 27 | p32750-rs7967521 | |
| 28 | p32777-rs11057841 | |
| 29 | p32860-rs7967406 | |
| 30 | p33531-rs11057838 | |
| 31 | p36094-rs11608336 | |
| 32 | p36361-rs4765178 | |
| 33 | p36908-rs10846745 | |
| 34 | p37095-rs10846744 | |
| 35 | p41632-rs6488943 | |
| 36 | p42467-rs11057830 | |
| 37 | **p45516-rs1902569** | |
| 38 | p49537-rs7305310 | |
| 39 | p49570delC-rs145376237 | |
| 40 | **p49690-rs4765615** | |
| 41 | p50151-rs2278986 | p48969-rs2343394 |
| 42 | p51888-rs7138304 | |
| 43 | p52096-rs10846739 | |
| 44 | p52556-rs11057820 | |
| 45 | p52610-rs10846738 | |
| 46 | p52956-rs77740046 | |
| 47 | p53359-rs112371713 | |

|  |  |  |
|---|---|---|
| 48 | p53790-rs4765614 |  |
| 49 | p54492-rs61762481 |  |
| 50 | p55923-rs838900 |  |
| 51 | **p55963-rs7134858** |  |
| 52 | p57107-rs5892 (p.Phe301Phe) |  |
| 53 | p57508-rs71458866 | p62615-rs7138386 |
| 54 | p57592-rs838903 |  |
| 55 | p58514-rs838905 | p56845-rs838902 |
| 56 | p58664-rs865716 |  |
| 57 | p61872-rs838909 |  |
| 58 | p62140-rs838910 |  |
| 59 | p62409-rs838911 |  |
| 60 | **p63483-rs838912** |  |
| 61 | **p64772-rs5888 (p.Ala350Ala)** |  |
| 62 | p64923-rs838915 |  |
| 63 | p65999-rs12819677 | p60255-rs3782287 |
| 64 | p67439-rs961170 |  |
| 65 | p67700-rs1726374 |  |
| 66 | p69013-rs7135117 | p69995delC-rs5801571 |
| 67 | p69699-rs10396210 |  |
| 68 | p71867-rs7954022 |  |
| 69 | p72197-rs838861 |  |
| 70 | p72777-rs838862 |  |
| 71 | p75766-rs838866 |  |
| 72 | p75778-rs7301120 |  |
| 73 | p76757-rs9919713 |  |
| 74 | p77842-rs2272310 |  |
| 75 | p78402-rs838898 |  |
| 76 | p78430-rs838897 |  |
| 77 | p78747-rs2293440 |  |
| 78 | **p79721-rs838896** |  |
| 79 | **p79828-rs838895** | p80045-rs838893 |
| 80 | p83547-rs838887 |  |
| 81 | **p83884-rs701106** |  |
| 82 | p86276-rs747155 |  |
| 83 | p86481-rs701103 (p.Gly499Arg, isoform 2) |  |
| 84 | p87011-rs58032386 |  |
| 85 | p87723-rs838881 |  |
| 86 | p87749-rs76465225 |  |
| 87 | p87927-rs838880 |  |

del, deletion; MAF, minor allele frequency; SNP, single nucleotide polymorphism.

Each SNP is shown as “SNP Name-SNP IDs (amino acid change)”; see Table B6 for details for each SNP.

SNP names and chromosome 12 positions were based on the RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics).

SNP IDs were based on dbSNP build 139: GRCh37.10.

SNPs with nominally significant evidence of single-site association with either high-density lipoprotein cholesterol or apolipoprotein A-I (*P* <0.05; Tables B9 and B10) observed in the current study are shown in **bold**.

## Table B8. Covariates used for lipid variables in statistical analyses.

| | Lipid Traits | | | | |
|---|---|---|---|---|---|
| **Covariates** | **HDL-C** | **LDL-C** | **TG** | **ApoA-I** | **ApoB** |
| **Sex (M/F)** | X | X | X | X | |
| **Age, years** | X | | X | X | |
| **Body mass index, kg/m$^2$** | | X | | | X |
| **Waist, cm** | X | | X | | |
| **Current smoking (yes/no)** | X | X | | | |
| **Jobmin, min** | X | X | X | | |
| **Staff (junior/senior)** | | X | | | X |

ApoA-I, apolipoprotein A-I; ApoB, apolipoprotein B; HDL-C, high-density lipoprotein cholesterol; LDL-C, low-density lipoprotein cholesterol; TG, triglycerides.
All phenotypic values were Box-Cox transformed prior to statistical analyses.
“X” represents covariates that were used for adjustment in statistical analyses.
Body mass index (BMI), weight (kg)/[height (m)]$^2$.
Jobmin, minutes of walking or biking to work each day.
Staff, occupational status: junior, non-professional staff with salary grades 1-6; and senior, professional and administrative staff with salary grades 7-16.
Waist, waist measurement at the narrowest point or at the umbilicus if the narrowest point was undetermined.

## Table B9. Single-site association results of 136 *SCARB1* genotyped variants for high-density lipoprotein cholesterol (HDL-C).

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | RegDB Score[d] | MA, MAF | Genotype | Genotype Count | Adjusted Mean (mg/dL) | SD (mg/dL) | β | SE | *P* | FDR[e] |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **MAF ≥5%** | | | | | | | | | | | | | | |
| p1265 | rs2070242 | 125348255 | Exon 1 | Ser4Ser | 2b | T, 0.1284 | CC/CT/TT | 541/163/10 | 48.38/46.08/49.75 | 12.44/13.11/12.75 | -0.5040 | 0.2906 | 0.0833 | 0.453 |
| p4072 | rs7139401 | 125345448 | Intron 1 | | 3b | C, 0.4386 | TT/TC/CC | 232/343/147 | 47.46/48.27/47.73 | 13.65/12.32/11.97 | 0.0877 | 0.1905 | 0.6453 | 0.820 |
| p5055 | rs11057869 | 125344465 | Intron 1 | | 7 | A, 0.0740 | AA/GA/GG | 7/96/638 | 45.72/48.5/47.75 | 12.97/12.78/12.69 | 0.0976 | 0.3538 | 0.7828 | 0.902 |
| p6600 | rs12831105 | 125342920 | Intron 1 | | 7 | T, 0.1188 | CC/CT/TT | 566/151/11 | 47.51/49.2/47.82 | 12.24/14.20/12.34 | 0.3570 | 0.2933 | 0.2239 | 0.683 |
| p10292 | rs4765181 | 125339228 | Intron 1 | | 5 | T, 0.2490 | TT/GT/GG | 44/285/417 | 46.42/48.72/47.39 | 12.15/12.86/12.52 | 0.1348 | 0.2201 | 0.5405 | 0.820 |
| p10991 | rs10773112 | 125338529 | Intron 1 | | 4 | A, 0.3534 | GG/GA/AA | 303/341/89 | 47.35/48.09/48.59 | 13.39/12.02/12.81 | 0.2146 | 0.2010 | 0.2860 | 0.683 |
| p13570 | rs11057864 | 125335950 | Intron 1 | | 4 | T, 0.1180 | TT/GT/GG | 14/147/581 | 49.3/47.24/47.89 | 9.25/11.91/12.98 | -0.0393 | 0.2886 | 0.8917 | 0.962 |
| p16565 | rs10773111 | 125332955 | Intron 1 | | 6 | A, 0.1928 | GG/GA/AA | 481/239/24 | 47.65/48.23/47.55 | 12.70/12.62/13.49 | 0.1089 | 0.2436 | 0.6550 | 0.820 |
| p20207 | rs11057853 | 125329313 | Intron 1 | | 5 | A, 0.4484 | AA/GA/GG | 150/360/228 | 48.27/48.84/45.85 | 12.70/13.23/11.74 | 0.4082 | 0.1925 | **0.0343** | 0.424 |
| p20694 | rs11057852 | 125328826 | Intron 1 | | 7 | A, 0.1050 | GG/GA/AA | 581/130/11 | 48.04/47.5/49.33 | 12.91/11.75/9.83 | -0.0308 | 0.3061 | 0.9198 | 0.962 |
| p20741 | rs11057851 | 125328779 | Intron 1 | | 5 | T, 0.3237 | TT/CT/CC | 72/334/328 | 43.9/47.68/48.86 | 11.05/13.00/12.57 | -0.5924 | 0.2067 | **0.0043** | **0.146** |
| p21145 | rs3924313 | 125328375 | Intron 1 | | 6 | T, 0.1772 | CC/CT/TT | 503/219/22 | 47.6/48.13/48.4 | 12.50/13.00/9.98 | 0.1488 | 0.2489 | 0.5502 | 0.820 |
| p22116 | rs12370382 | 125327404 | Intron 1 | | 1f | A, 0.0645 | GG/GA/AA | 643/86/3 | 47.57/49.06/35.39 | 12.67/13.28/6.62 | 0.1369 | 0.3965 | 0.7300 | 0.871 |
| p22168 | rs7137797 | 125327352 | Intron 1 | | 2b | C, 0.3977 | CC/TC/TT | 113/355/259 | 48.48/48.04/47.1 | 13.14/12.49/12.77 | 0.2153 | 0.1976 | 0.2763 | 0.683 |
| p22331 | rs6488944 | 125327189 | Intron 1 | | 4 | G, 0.1474 | TT/TG/GG | 522/188/12 | 47.54/48.91/47.47 | 12.66/12.99/12.80 | 0.3091 | 0.2778 | 0.2661 | 0.683 |
| p22675 | rs12425134 | 125326845 | Intron 1 | | 4 | T, 0.0526 | TT/GT/GG | 2/75/662 | 54.65/49.92/47.6 | 4.88/11.66/12.79 | 0.7556 | 0.4221 | 0.0738 | 0.453 |
| p28137 | rs12229555 | 125321383 | Intron 1 | | 7 | G, 0.3896 | GG/AG/AA | 99/373/257 | 46.88/47.29/49.16 | 12.96/12.60/12.64 | -0.3976 | 0.2025 | 0.0500 | 0.447 |
| p28692 | rs4765622 | 125320828 | Intron 1 | | 5 | T, 0.2565 | TT/CT/CC | 43/289/403 | 47.61/48.48/47.36 | 13.61/12.50/12.84 | 0.1962 | 0.2242 | 0.3817 | 0.763 |
| p28957 | rs11057844 | 125320563 | Intron 1 | | 5 | A, 0.2362 | GG/GA/AA | 428/263/40 | 47.34/47.87/50.96 | 13.11/12.40/10.13 | 0.3671 | 0.2278 | 0.1075 | 0.515 |
| p29749 | rs10846751 | 125319771 | Intron 1 | | 7 | T, 0.4492 | TT/CT/CC | 142/365/210 | 46.42/47.81/48.73 | 13.57/12.59/12.27 | -0.3558 | 0.1961 | 0.0701 | 0.453 |
| p31072 | rs10846749 | 125318448 | Intron 1 | | 4 | G, 0.4461 | CC/CG/GG | 222/366/144 | 48.83/47.61/46.39 | 12.22/12.86/13.04 | -0.3792 | 0.1940 | 0.0510 | 0.447 |
| p31938 | rs10744182 | 125317582 | Intron 1 | | 5 | A, 0.1837 | AA/GA/GG | 25/221/487 | 45.31/47.42/47.94 | 16.27/13.13/12.29 | -0.2885 | 0.2474 | 0.2438 | 0.683 |
| p32129 | rs10773107 | 125317391 | Intron 1 | | 7 | T, 0.1009 | TT/GT/GG | 9/132/593 | 47/46.79/48.03 | 8.10/12.92/12.72 | -0.3186 | 0.3141 | 0.3107 | 0.693 |
| p32273 | rs12580803 | 125317247 | Intron 1 | | 5 | C, 0.1006 | TT/TC/CC | 619/114/18 | 47.7/47.52/51.35 | 12.85/11.79/13.59 | 0.2010 | 0.2968 | 0.4984 | 0.820 |
| p32290 | rs10744181 | 125317230 | Intron 1 | | 5 | C, 0.1238 | TT/TC/CC | 547/141/18 | 47.89/48.46/42.63 | 12.60/13.86/9.02 | -0.1852 | 0.2851 | 0.5162 | 0.820 |
| p32395 | rs12581963 | 125317125 | Intron 1 | | 5 | T, 0.1314 | TT/CT/CC | 11/179/557 | 47.47/46.69/48.1 | 8.81/11.70/12.94 | -0.3173 | 0.2808 | 0.2589 | 0.683 |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p32750 | rs7967521 | 125316770 | Intron 1 | | 7 | G, 0.3425 | GG/AG/AA | 89/306/310 | 45.26/48.42/47.89 | 11.20/12.62/13.26 | -0.2137 | 0.2034 | 0.2936 | 0.683 |
| p32777 | rs11057841 | 125316743 | Intron 1 | | 7 | A, 0.2805 | GG/GA/AA | 367/307/50 | 47.82/48.49/44.86 | 12.82/12.79/10.19 | -0.1217 | 0.2187 | 0.5781 | 0.820 |
| p32860 | rs7967406 | 125316660 | Intron 1 | | 6 | C, 0.0991 | CC/AC/AA | 9/131/608 | 46.8/48.71/47.54 | 8.12/12.43/12.81 | 0.2647 | 0.3141 | 0.3996 | 0.788 |
| p33531 | rs11057838 | 125315989 | Intron 1 | | 7 | A, 0.2278 | CC/CA/AA | 439/254/41 | 48.09/47.82/45.51 | 12.94/12.64/10.74 | -0.2064 | 0.2258 | 0.3609 | 0.763 |
| p36094 | rs11608336 | 125313426 | Intron 1 | | 4 | A, 0.1543 | GG/GA/AA | 521/195/15 | 47.49/48.96/48.39 | 12.47/13.43/10.99 | 0.3354 | 0.2678 | 0.2108 | 0.683 |
| p36461 | rs4765178 | 125313059 | Intron 1 | | 4 | T, 0.1671 | TT/CT/CC | 19/201/496 | 47.45/47.5/48.12 | 10.94/12.52/12.99 | -0.1369 | 0.2628 | 0.6024 | 0.820 |
| p36908 | rs10846745 | 125312612 | Intron 1 | | 4 | G, 0.3257 | GG/CG/CC | 76/334/329 | 44.67/49.15/47.23 | 10.95/13.05/12.30 | -0.0384 | 0.2045 | 0.8511 | 0.949 |
| p37095 | rs10846744 | 125312425 | Intron 1 | | 4 | G, 0.3056 | CC/CG/GG | 348/340/57 | 47.72/47.75/48.9 | 13.10/11.99/13.09 | 0.1168 | 0.2125 | 0.5829 | 0.820 |
| p41632 | rs6488943 | 125307888 | Intron 1 | | 5 | C, 0.2954 | CC/AC/AA | 50/309/337 | 45.63/48.2/48.34 | 9.68/13.16/12.65 | -0.2195 | 0.2226 | 0.3244 | 0.704 |
| p42467 | rs11057830 | 125307053 | Intron 1 | | 7 | T, 0.1523 | TT/CT/CC | 13/201/523 | 40.37/48.03/47.91 | 15.96/13.41/12.33 | -0.2810 | 0.2718 | 0.3015 | 0.683 |
| p45516 | rs1902569 | 125304004 | Intron 1 | | 5 | A, 0.1544 | AA/GA/GG | 18/190/519 | 49.5/49.31/47.28 | 12.57/11.81/12.95 | 0.5447 | 0.2629 | **0.0386** | 0.438 |
| p48969 | rs2343394 | 125300551 | Intron 2 | | 5 | T, 0.1898 | TT/CT/CC | 32/225/491 | 47.56/48.89/47.23 | 11.15/12.64/12.73 | 0.3165 | 0.2352 | 0.1788 | 0.624 |
| p49537 | rs7305310 | 125299983 | Intron 2 | | 5 | T, 0.1007 | CC/CT/TT | 595/117/16 | 48.06/47.04/45.04 | 12.87/12.58/9.83 | -0.3396 | 0.2991 | 0.2566 | 0.683 |
| p49570 delC | rs145376237 | 125299950 | Intron 2 | | 5 | delC, 0.2276 | DD/WD/WW | 36/260/432 | 44.33/49.8/46.89 | 11.46/13.26/12.43 | 0.3121 | 0.2311 | 0.1773 | 0.624 |
| p49690 | rs4765615 | 125299830 | Intron 2 | | 5 | A, 0.4426 | AA/GA/GG | 156/318/244 | 45.87/48/49.03 | 13.19/12.44/13.01 | -0.4646 | 0.1866 | **0.0130** | 0.253 |
| p50151 | rs2278986 | 125299369 | Intron 3 | | 5 | C, 0.1933 | CC/TC/TT | 33/225/484 | 49.64/48.72/47.21 | 11.26/12.41/12.86 | 0.4333 | 0.2350 | 0.0656 | 0.453 |
| p51888 | rs7138304 | 125297632 | Intron 4 | | 2b | T, 0.1079 | TT/CT/CC | 16/129/589 | 45.08/48.13/47.83 | 9.22/12.36/12.75 | -0.0610 | 0.2898 | 0.8334 | 0.937 |
| p52096 | rs10846739 | 125297424 | Intron 4 | | 3a | G, 0.4693 | GG/AG/AA | 169/343/217 | 46.18/49.5/46.33 | 13.08/12.63/12.17 | 0.0195 | 0.1868 | 0.9168 | 0.962 |
| p52556 | rs11057820 | 125296964 | Intron 4 | | 5 | A, 0.1000 | AA/GA/GG | 12/124/610 | 40.36/48.71/47.69 | 11.34/13.80/12.49 | -0.1513 | 0.3081 | 0.6235 | 0.820 |
| p52610 | rs10846738 | 125296910 | Intron 4 | | 4 | T, 0.1349 | TT/CT/CC | 12/172/545 | 46.62/48.16/47.93 | 12.58/13.45/12.36 | -0.0102 | 0.2804 | 0.9710 | 0.985 |
| p52956 | rs77740046 | 125296564 | Intron 4 | | 5 | T, 0.0546 | CC/CT/TT | 651/75/3 | 47.69/48.97/46.21 | 12.89/11.66/13.34 | 0.3300 | 0.4168 | 0.4288 | 0.802 |
| p53359 | rs112371713 | 125296161 | Intron 5 | | 5 | A, 0.1243 | AA/GA/GG | 9/160/549 | 44.06/49.59/47.4 | 13.40/12.95/12.69 | 0.4193 | 0.3018 | 0.1651 | 0.624 |
| p53790 | rs4765614 | 125295730 | Intron 5 | | 5 | A, 0.2653 | GG/GA/AA | 399/276/58 | 48.17/48.05/44.21 | 13.17/12.42/10.42 | -0.3281 | 0.2118 | 0.1218 | 0.537 |
| p54492 | rs61762481 | 125295028 | Intron 5 | | 4 | A, 0.1005 | AA/GA/GG | 8/137/607 | 47.58/47.31/47.92 | 8.15/12.76/12.74 | -0.1522 | 0.3119 | 0.6257 | 0.820 |
| p55923 | rs838900 | 125293597 | Intron 6 | | 7 | A, 0.3921 | AA/GA/GG | 113/345/275 | 47.05/49.45/46.18 | 13.85/13.31/11.20 | 0.2787 | 0.1957 | 0.1549 | 0.619 |
| p55963 | rs7134858 | 125293557 | Intron 6 | | 6 | T, 0.1560 | TT/CT/CC | 24/184/532 | 50.7/48.34/47.23 | 7.08/11.91/12.90 | 0.4418 | 0.2520 | 0.0799 | 0.453 |
| p56845 | rs838902 | 125292675 | Intron 6 | | 5 | G, 0.4249 | AA/AG/GG | 249/347/141 | 48.01/47.88/47.42 | 12.84/12.60/12.79 | -0.0786 | 0.1905 | 0.6801 | 0.834 |
| p57107 | rs5892 | 125292413 | Exon 7 | Phe301Phe | 4 | T, 0.0589 | CC/CT/TT | 656/85/1 | 47.84/47.53/54.22 | 12.92/11.23/NA | 0.0079 | 0.4138 | 0.9848 | 0.985 |
| p57508 | rs71458866 | 125292012 | Intron 7 | | 4 | A, 0.1130 | AA/GA/GG | 13/144/594 | 47.5/47.11/47.93 | 9.18/12.92/12.68 | -0.1926 | 0.2914 | 0.5089 | 0.820 |
| p57592 | rs838903 | 125291928 | Intron 7 | | 4 | A, 0.3763 | AA/GA/GG | 111/334/290 | 47.18/47.64/48.29 | 11.62/13.06/12.86 | -0.1598 | 0.1959 | 0.4149 | 0.802 |
| p58514 | rs838905 | 125291006 | Intron 7 | | 4 | C, 0.4329 | CC/TC/TT | 146/350/243 | 47.34/47.97/48.02 | 12.68/12.56/13.04 | -0.0855 | 0.1905 | 0.6536 | 0.820 |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p58664 | rs865716 | 125290856 | Intron 7 | | 5 | T, 0.2708 | AA/AT/TT | 395/268/62 | 48.35/46.87/49.05 | 13.05/12.47/10.76 | -0.0965 | 0.2094 | 0.6449 | 0.820 |
| p60255 | rs3782287 | 125289265 | Intron 7 | | 5 | T, 0.2831 | CC/CT/TT | 378/312/51 | 46.89/48.96/46.64 | 12.02/13.23/14.24 | 0.2555 | 0.2197 | 0.2453 | 0.683 |
| p61872 | rs838909 | 125287648 | Intron 7 | | 4 | T, 0.2199 | CC/CT/TT | 449/249/36 | 47.59/48.12/48.15 | 12.60/13.16/10.94 | 0.1223 | 0.2297 | 0.5945 | 0.820 |
| p62140 | rs838910 | 125287380 | Intron 7 | | 5 | T, 0.3047 | GG/GT/TT | 355/304/70 | 47.6/48.51/45.61 | 12.61/12.92/12.07 | -0.0759 | 0.2071 | 0.7143 | 0.860 |
| p62409 | rs838911 | 125287111 | Intron 7 | | 5 | T, 0.4211 | CC/CT/TT | 247/347/133 | 48.21/48.12/46.87 | 13.10/12.87/11.68 | -0.1556 | 0.1934 | 0.4213 | 0.802 |
| p62615 | rs7138386 | 125286905 | Intron 7 | | 5 | C, 0.1137 | TT/TC/CC | 568/138/12 | 48.18/46.68/50.88 | 12.66/12.76/9.42 | -0.2083 | 0.2982 | 0.4851 | 0.820 |
| p63483 | rs838912 | 125286037 | Intron 7 | | 7 | A, 0.0867 | AA/GA/GG | 6/117/615 | 47.88/48.51/47.79 | 8.05/12.85/12.52 | 0.1814 | 0.3311 | 0.5840 | 0.820 |
| p64772 | rs5888 | 125284748 | Exon 8 | Ala350Ala | 3a | T, 0.0961 | CC/CT/TT | 605/129/8 | 47.6/48.54/52.11 | 12.65/13.16/10.27 | 0.3620 | 0.3186 | 0.2561 | 0.683 |
| p64923 | rs838915 | 125284597 | Intron 8 | | 5 | A, 0.1435 | AA/CA/CC | 19/177/539 | 45.25/48.03/47.87 | 9.15/12.97/12.72 | -0.0858 | 0.2654 | 0.7466 | 0.883 |
| p65999 | rs12819677 | 125283521 | Intron 8 | | 6 | A, 0.2813 | GG/GA/AA | 371/310/50 | 46.78/49.09/47.82 | 12.14/13.01/13.90 | 0.4021 | 0.2192 | 0.0670 | 0.453 |
| p67439 | rs961170 | 125282081 | Intron 8 | | 4 | A, 0.0893 | AA/GA/GG | 11/108/610 | 45.83/48.01/47.82 | 17.60/12.30/12.70 | -0.0481 | 0.3220 | 0.8812 | 0.962 |
| p67700 | rs1726374 | 125281820 | Intron 8 | | 7 | A, 0.1933 | GG/GA/AA | 491/221/35 | 47.61/47.44/51.33 | 12.81/12.01/14.89 | 0.2289 | 0.2330 | 0.3262 | 0.704 |
| p69013 | rs7135117 | 125280507 | Intron 8 | | 7 | G, 0.2901 | GG/AG/AA | 75/275/377 | 48.2/48.1/47.63 | 12.45/12.42/13.01 | 0.1133 | 0.2024 | 0.5758 | 0.820 |
| p69699 | rs10396210 | 125279821 | Intron 8-splice site | | 4 | A, 0.1511 | AA/GA/GG | 17/186/523 | 46.35/46.27/48.29 | 10.51/11.82/13.06 | -0.4778 | 0.2675 | 0.0745 | 0.453 |
| p69995 delC | rs5801571 | 125279525 | Intron 9 | | 5 | delC, 0.2761 | DD/WD/WW | 63/281/386 | 47.7/47.72/47.45 | 13.42/12.34/13.04 | 0.0614 | 0.2123 | 0.7724 | 0.898 |
| p71867 | rs7954022 | 125277653 | Intron 9 | | 5 | T, 0.1323 | TT/CT/CC | 11/172/552 | 45/49.47/47.43 | 12.10/12.93/12.59 | 0.3876 | 0.2864 | 0.1764 | 0.624 |
| p72197 | rs838861 | 125277323 | Intron 9 | | 7 | G, 0.3777 | AA/AG/GG | 300/308/123 | 48.56/47.21/47.97 | 13.20/12.13/13.06 | -0.1415 | 0.1883 | 0.4527 | 0.810 |
| p72777 | rs838862 | 125276743 | Intron 9 | | 5 | T, 0.0887 | CC/CT/TT | 607/115/7 | 47.9/47.97/51.52 | 12.46/13.53/18.41 | 0.1013 | 0.3334 | 0.7613 | 0.893 |
| p75766 | rs838866 | 125273754 | Intron 9 | | 6 | C, 0.2116 | TT/TC/CC | 457/239/38 | 48.02/47.68/47.04 | 12.90/12.55/10.89 | -0.1058 | 0.2276 | 0.6423 | 0.820 |
| p75778 | rs7301120 | 125273742 | Intron 9 | | 6 | T, 0.1135 | TT/CT/CC | 9/147/563 | 50.67/46.79/48.17 | 7.48/12.53/12.82 | -0.2367 | 0.3041 | 0.4366 | 0.802 |
| p76757 | rs9919713 | 125272763 | Intron 9 | | 6 | T, 0.4390 | AA/AT/TT | 235/347/151 | 48.16/47.8/47.14 | 13.78/12.06/12.19 | -0.1264 | 0.1892 | 0.5044 | 0.820 |
| p77251 | rs34339961 | 125272269 | Intron 9 | | 6 | T, 0.1177 | AA/AT/TT | 561/151/11 | 47.91/46.76/50.68 | 12.87/12.10/8.13 | -0.1372 | 0.2955 | 0.6426 | 0.820 |
| p77842 | rs2272310 | 125271678 | Intron 10 | | 5 | A, 0.0807 | AA/GA/GG | 5/110/630 | 51.11/48.16/47.65 | 12.21/13.23/12.64 | 0.2011 | 0.3494 | 0.5651 | 0.820 |
| p78402 | rs838898 | 125271118 | Intron 10 | | 5 | A, 0.0714 | AA/GA/GG | 7/86/594 | 45.47/48/48.01 | 7.94/12.21/13.05 | -0.0602 | 0.3738 | 0.8720 | 0.962 |
| p78430 | rs838897 | 125271090 | Intron 10 | | 5 | G, 0.3830 | GG/CG/CC | 125/308/291 | 46.86/48.36/47.33 | 12.37/12.40/12.86 | 0.0070 | 0.1877 | 0.9704 | 0.985 |
| p78747 | rs2293440 | 125270773 | Intron 11 | | 5 | C, 0.4112 | CC/TC/TT | 128/342/252 | 46.7/47.72/48.12 | 10.50/12.59/13.48 | -0.1684 | 0.1919 | 0.3806 | 0.763 |
| p79721 | rs838896 | 125269799 | Intron 11 | | 5 | C, 0.3104 | GG/GC/CC | 349/319/73 | 47.39/47.6/50.86 | 13.07/12.12/12.38 | 0.3565 | 0.2045 | 0.0817 | 0.453 |
| p79828 | rs838895 | 125269692 | Intron 11 | | 5 | G, 0.3171 | GG/CG/CC | 74/322/337 | 50.65/48.22/46.95 | 12.74/12.27/13.10 | 0.4961 | 0.2059 | **0.0162** | 0.276 |
| p80045 | rs838893 | 125269475 | Intron 11 | | 5 | A, 0.3244 | GG/GA/AA | 335/325/81 | 47.17/48.1/49.33 | 12.98/12.45/12.54 | 0.3127 | 0.2022 | 0.1224 | 0.537 |
| p83547 | rs838887 | 125265973 | Intron 12 | | 5 | G, 0.4564 | CC/CG/GG | 225/357/154 | 46.99/48.49/47.62 | 12.57/12.51/13.34 | 0.1202 | 0.1898 | 0.5267 | 0.820 |
| p83884 | rs701106 | 125265636 | Intron 12 | | 5 | T, 0.2597 | TT/CT/CC | 49/289/405 | 50.45/47.79/47.56 | 13.58/12.76/12.53 | 0.2471 | 0.2192 | 0.2601 | 0.683 |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p86276 | rs747155 | 125263244 | Intron 12 | | 2b | T, 0.1495 | TT/CT/CC | 17/187/533 | 48.37/48.5/47.47 | 13.90/11.86/12.96 | 0.2793 | 0.2681 | 0.2980 | 0.683 |
| p86481 | rs701103 | 125263039 | Exon 13-3' UTR | Gly499Arg (isoform 2) | 5 | A, 0.2451 | AA/GA/GG | 50/259/424 | 46.92/48.83/47.38 | 13.72/12.39/12.59 | 0.1642 | 0.2169 | 0.4492 | 0.810 |
| p87011 | rs58032386 | 125262509 | Exon 13-3' UTR | | 2a | T, 0.1417 | CC/CT/TT | 544/183/14 | 47.8/48.03/47.93 | 12.88/11.70/16.17 | 0.0575 | 0.2733 | 0.8333 | 0.937 |
| p87723 | rs838881 | 125261797 | 3' flanking | | 6 | T, 0.3183 | TT/CT/CC | 63/341/332 | 47.01/48.22/47.16 | 12.74/12.21/13.05 | 0.1390 | 0.2132 | 0.5146 | 0.820 |
| p87749 | rs76465225 | 125261771 | 3' flanking | | 7 | A, 0.0844 | AA/GA/GG | 7/109/620 | 43.03/47.81/47.93 | 19.91/12.52/12.66 | -0.1992 | 0.3402 | 0.5583 | 0.820 |
| p87927 | rs838880 | 125261593 | 3' flanking | | 5 | A, 0.2414 | AA/GA/GG | 39/275/418 | 44.74/48.67/47.52 | 12.40/12.15/13.23 | 0.0198 | 0.2302 | 0.9314 | 0.967 |
| **MAF between 1-5%** | | | | | | | | | | | | | | |
| p1316 | rs10396208 | 125348204 | Exon 1 | Cys21Cys | 2b | T, 0.0476 | CC/CT/TT | 652/58/4 | 47.77/49.2/47.27 | 12.64/12.69/18.13 | 0.2987 | 0.4428 | 0.5001 | 0.820 |
| p1419 | rs201717369 | 125348101 | Intron 1 | | 4 | A, 0.0121 | AA/GA/GG | 1/14/707 | 55.72/48.94/47.8 | NA/15.28/12.67 | 0.4892 | 0.8717 | 0.5748 | 0.820 |
| p7650 | rs11615630 | 125341870 | Intron 1 | | 5 | A, 0.0436 | GG/GA | 685/67 | 47.63/49.16 | 12.20/16.83 | 0.3382 | 0.4681 | 0.4702 | 0.820 |
| p45627 | rs12297372 | 125303893 | Intron 1 | | 5 | G, 0.0487 | GG/AG/AA | 1/68/659 | 30.17/48.3/47.89 | NA/12.68/12.74 | -0.0483 | 0.4556 | 0.9156 | 0.962 |
| p46964 | rs114061302 | 125302556 | Intron 1 | | 4 | A, 0.0388 | AA/GA/GG | 1/55/688 | 34.33/48.54/47.73 | NA/13.75/12.65 | 0.0526 | 0.4975 | 0.9158 | 0.962 |
| p50118 | rs58710319 | 125299402 | Intron 3 | | 5 | T, 0.0208 | CT/CC | 31/711 | 50.42/47.73 | 13.43/12.65 | 0.7885 | 0.6704 | 0.2399 | 0.683 |
| p50380 | rs141748317 | 125299140 | Intron 3 | | 2b | G, 0.0112 | AA/AG | 723/15 | 47.81/48.13 | 12.68/15.76 | -0.0273 | 0.9564 | 0.9772 | 0.985 |
| p50489 | rs61320152 | 125299031 | Intron 3 | | 4 | T, 0.0257 | GG/GT | 699/38 | 47.82/48.65 | 12.69/13.60 | 0.2390 | 0.6110 | 0.6958 | 0.845 |
| p54445 | rs60910935 | 125295075 | Intron 5 | | 4 | G, 0.0418 | AA/AG/GG | 650/56/2 | 47.98/46.86/53.78 | 13.00/10.86/1.84 | -0.1247 | 0.4830 | 0.7963 | 0.910 |
| p54475 | rs60227139 | 125295045 | Intron 5 | | 4 | T, 0.0437 | CC/CT/TT | 670/61/2 | 47.84/47.01/53.7 | 12.92/10.81/1.84 | -0.0602 | 0.4628 | 0.8966 | 0.962 |
| p77682 | rs150082885 | 125271838 | Intron 10 | | 5 | G, 0.0106 | AA/AG/GG | 716/13/1 | 47.88/48.15/31.47 | 12.76/13.91/NA | -0.5377 | 0.9010 | 0.5509 | 0.820 |
| p78791 | rs75289200 | 125270729 | Intron 11 | | 5 | C, 0.0321 | TC/TT | 46/679 | 50.26/47.65 | 14.67/12.57 | 0.7037 | 0.5581 | 0.2078 | 0.683 |
| p82019 | rs838890 | 125267501 | Intron 11 | | 5 | T, 0.0320 | CC/CT/TT | 683/42/2 | 48.07/44.74/39.26 | 12.91/10.83/17.25 | -1.0051 | 0.5373 | 0.0618 | 0.453 |
| p82340 | rs77483223 | 125267180 | Intron 12 | | 5 | A, 0.0231 | GA/GG | 35/699 | 44.56/47.96 | 11.90/12.76 | -1.0458 | 0.6373 | 0.1012 | 0.510 |
| p82434 | rs838889 | 125267086 | Intron 12 | | 5 | C, 0.0315 | CC/TC/TT | 2/42/695 | 39.33/44.57/48.04 | 17.25/10.84/12.85 | -1.0389 | 0.5352 | 0.0526 | 0.447 |
| p86245 | rs188375019 | 125263275 | Intron 12 | | 4 | T, 0.0341 | CC/CT | 690/50 | 47.7/50.2 | 12.65/12.86 | 0.7447 | 0.5344 | 0.1639 | 0.624 |
| p86316 | rs701104 | 125263204 | Intron 12 | | 4 | T, 0.0487 | TT/GT/GG | 2/66/643 | 39.94/45.01/48.22 | 7.78/11.60/12.86 | -0.9838 | 0.4484 | **0.0286** | 0.389 |
| p86967 | rs187492239 | 125262553 | Exon 13-3' UTR | | 4 | G, 0.0355 | AA/AG | 686/52 | 47.68/50.25 | 12.71/12.51 | 0.7743 | 0.5256 | 0.1412 | 0.582 |
| p87611 | rs190688220 | 125261909 | 3' flanking | | 4 | T, 0.0316 | CC/CT | 691/46 | 47.66/50.44 | 12.72/12.61 | 0.8329 | 0.5573 | 0.1355 | 0.576 |
| p87681 | rs838883 | 125261839 | 3' flanking | | 5 | A, 0.0459 | AA/GA/GG | 1/65/646 | 45.6/44.85/48.19 | NA/11.43/12.90 | -0.9433 | 0.4647 | **0.0427** | 0.447 |
| **MAF ≤1%** | | | | | | | | | | | | | | |
| p1048 insC (1048_1049) | | 125348472 | Exon 1-5' UTR | | 2a | insC, 0.0079 | WI/WW | 12/723 | 45.31/47.92 | 15.77/12.65 | -0.9389 | 1.0655 | 0.3785 | 0.763 |
| p49759 | rs146272788 | 125299761 | Intron 2 | | 5 | T, 0.0020 | CC/CT | 725/3 | 47.84/56.6 | 12.78/13.38 | 2.5988 | 2.1256 | 0.2219 | 0.683 |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p49978 | rs5891 | 125299542 | Exon 3 | Val135Ile | 5 | A, 0.0058 | GA/GG | 9/743 | 52.42/47.7 | 18.19/12.61 | 1.3374 | 1.2347 | 0.2791 | 0.683 |
| p50024 | rs368880622 | 125299496 | Intron 3 | | 5 | T, 0.0026 | GG/GT | 737/3 | 47.83/53.32 | 12.72/12.83 | 1.6506 | 2.1188 | 0.4362 | 0.802 |
| p50954 | | 125298566 | Intron 4 | | 5 | C, 0.0007 | TC/TT | 1/735 | 63.83/47.81 | NA/12.71 | 4.5639 | 3.6627 | 0.2131 | 0.683 |
| p52919 | | 125296601 | Intron 4 | | 5 | T, 0.0013 | GG/GT | 734/2 | 47.87/24.62 | 12.71/9.26 | -7.4063 | 2.5863 | **0.0043** | **0.146** |
| p52995 | rs113910315 | 125296525 | Intron 4-splice site | | 5 | G, 0.0020 | TG/TT | 3/740 | 43.06/47.83 | 12.96/12.72 | -1.4175 | 2.1192 | 0.5038 | 0.820 |
| p53372 | rs115604379 | 125296148 | Intron 5 | | 5 | T, 0.0066 | CC/CT | 729/10 | 47.68/58.2 | 12.64/13.03 | 3.0372 | 1.1642 | **0.0093** | 0.219 |
| p54611 | | 125294909 | Intron 5 | | 4 | C, 0.0007 | TC/TT | 1/742 | 19.59/47.86 | NA/12.68 | -9.5243 | 3.6710 | **0.0097** | 0.219 |
| p54627 | | 125294893 | Intron 5 | | 4 | C, 0.0020 | GC/GG | 3/733 | 50.9/47.79 | 15.45/12.75 | 0.9473 | 2.1330 | 0.6571 | 0.820 |
| p54856 | | 125294664 | Intron 6 | | 4 | T, 0.0007 | CC/CT | 742/1 | 47.85/21.48 | 12.70/NA | -8.4305 | 3.6579 | **0.0215** | 0.324 |
| p57004 | rs187562853 | 125292516 | Intron 6 | | 4 | A, 0.0098 | GG/GA | 721/15 | 47.61/53.57 | 12.63/18.00 | 1.6474 | 0.9619 | 0.0872 | 0.456 |
| p77181 | rs146246031 | 125272339 | Intron 9 | | 7 | C, 0.0053 | TC/TT | 6/731 | 45.71/47.91 | 12.57/12.72 | -0.6173 | 1.5007 | 0.6809 | 0.834 |
| p77381 | rs138499966 | 125272139 | Intron 9 | | 6 | C, 0.0046 | TC/TT | 7/735 | 53.23/47.76 | 17.65/12.68 | 1.5523 | 1.3921 | 0.2652 | 0.683 |
| p77620 | rs377124254 | 125271900 | Intron 10 | | 5 | A, 0.0007 | GA/GG | 1/735 | 90.2/47.77 | NA/12.67 | 11.5518 | 3.6514 | **0.0016** | **0.110** |
| p77704 | | 125271816 | Intron 10 | | 5 | A, 0.0040 | CA/CC | 5/726 | 42.75/47.86 | 9.16/12.79 | -1.4602 | 1.6591 | 0.3791 | 0.763 |
| p78255 | rs184052375 | 125271265 | Intron 10 | | 4 | G, 0.0072 | AA/AG | 732/11 | 47.75/52.05 | 12.64/17.17 | 1.1808 | 1.1134 | 0.2893 | 0.683 |
| p81863 | rs185445624 | 125267657 | Intron 11 | | 5 | A, 0.0020 | GA/GG | 3/739 | 45.05/47.83 | 20.36/12.71 | -0.9612 | 2.1241 | 0.6510 | 0.820 |
| p82264 | rs141545424 | 125267256 | Exon 12 | Gly501Gly | 5 | A, 0.0007 | CA/CC | 1/739 | 90.31/47.77 | NA/12.66 | 11.5850 | 3.6469 | **0.0016** | **0.110** |
| p82369 | rs75446635 | 125267151 | Intron 12 | | 5 | A, 0.0059 | GA/GG | 9/733 | 50.65/47.8 | 20.95/12.59 | 0.5896 | 1.2312 | 0.6322 | 0.820 |
| p87266 | rs150512235 | 125262254 | Exon 13-3' UTR[f] | | 4 | C, 0.0057 | TC/TT | 9/746 | 48.46/47.75 | 20.83/12.58 | -0.0325 | 1.2316 | 0.9789 | 0.985 |
| p87694 | | 125261826 | 3' flanking | | 5 | T, 0.0020 | CC/CT | 722/3 | 47.85/59.52 | 12.75/12.25 | 3.4021 | 2.1251 | 0.1098 | 0.515 |

del/D, deletion; FDR, false discovery rate; ins/I, insertion; MA, minor allele; MAF, minor allele frequency; NA, not analyzed; RegDB, RegulomeDB score; SD, standard deviation; SE, standard error; SNP, single nucleotide polymorphism; UTR, untranslated region; W, wild type allele on RefSeq for insertion and deletion variations.

All alleles on reverse strand. Splice site is defined as ± 20 bp from the start or end of an exon.

HDL-C values were Box-Cox transformed. Results were adjusted for covariates: sex, age, waist, current smoking (yes/no), and minutes of daily walking or biking to work (jobmin).

Nominally significant *P*-values ($P < 0.05$) and FDR values that passed the threshold (FDR <0.20) are shown in **bold**.

[a, c] Based on the RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics).

[b] dbSNP build 139: GRCh37.p10; 10 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH.(batch ID: SCARB1_AB).

[d] Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table B4 or can be seen at http://regulome.stanford.edu/help.

[e] FDR is referred to a *q*-value from Benjamini-Hochberg procedure.

[f] Close to the miRNA-145 seed binding site based on TargetScanHuman (version 6.2, http://www.targetscan.org/).

## Table B10. Single-site association results of 136 *SCARB1* genotyped variants for apolipoprotein A-I (ApoA-I).

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | RegDB Score[d] | MA, MAF | Genotype | Genotype Count | Adjusted Mean (mg/dL) | SD (mg/dL) | β | SE | *P* | FDR[e] |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **MAF ≥5%** | | | | | | | | | | | | | | |
| p1265 | rs2070242 | 125348255 | Exon 1 | Ser4Ser | 2b | T, 0.1284 | CC/CT/TT | 545/165/10 | 138.25/132.98/141.25 | 28.10/26.38/36.12 | -1.2535 | 0.7296 | 0.0862 | 0.558 |
| p4072 | rs7139401 | 125345448 | Intron 1 | | 3b | C, 0.4386 | TT/TC/CC | 236/350/148 | 136.26/138.43/135.18 | 27.17/28.38/27.51 | -0.0802 | 0.4732 | 0.8654 | 0.965 |
| p5055 | rs11057869 | 125344465 | Intron 1 | | 7 | A, 0.0740 | AA/GA/GG | 7/95/650 | 138.23/138.44/136.59 | 27.94/25.35/28.24 | 0.5638 | 0.8858 | 0.5247 | 0.849 |
| p6600 | rs12831105 | 125342920 | Intron 1 | | 7 | T, 0.1188 | CC/CT/TT | 571/152/10 | 137.08/135.15/135.11 | 27.64/27.93/25.69 | -0.5681 | 0.7373 | 0.4412 | 0.811 |
| p10292 | rs4765181 | 125339228 | Intron 1 | | 5 | T, 0.2490 | TT/GT/GG | 44/287/426 | 132.59/138.1/136.53 | 27.04/27.35/28.00 | -0.0286 | 0.5474 | 0.9583 | 0.993 |
| p10991 | rs10773112 | 125338529 | Intron 1 | | 4 | A, 0.3534 | GG/GA/AA | 309/339/90 | 135/138.69/135.87 | 28.55/26.69/27.19 | 0.4896 | 0.4950 | 0.3230 | 0.776 |
| p13570 | rs11057864 | 125335950 | Intron 1 | | 4 | T, 0.1180 | TT/GT/GG | 15/147/591 | 131.15/138.99/136.48 | 19.01/28.45/27.80 | 0.3006 | 0.7107 | 0.6725 | 0.905 |
| p16565 | rs10773111 | 125332955 | Intron 1 | | 6 | A, 0.1928 | GG/GA/AA | 486/246/23 | 135.99/138.39/134.71 | 27.30/28.56/28.10 | 0.4484 | 0.6077 | 0.4608 | 0.823 |
| p20207 | rs11057853 | 125329313 | Intron 1 | | 5 | A, 0.4484 | AA/GA/GG | 153/366/230 | 136.24/139.87/132.18 | 27.54/29.21/25.20 | 0.8422 | 0.4726 | 0.0751 | 0.554 |
| p20694 | rs11057852 | 125328826 | Intron 1 | | 7 | A, 0.1050 | GG/GA/AA | 587/129/11 | 136.59/138.27/135.18 | 27.73/28.21/20.66 | 0.3519 | 0.7640 | 0.6452 | 0.900 |
| p20741 | rs11057851 | 125328779 | Intron 1 | | 5 | T, 0.3237 | TT/CT/CC | 73/336/337 | 130.12/136.32/138.94 | 27.67/27.25/28.14 | -1.2331 | 0.5117 | **0.0162** | 0.319 |
| p21145 | rs3924313 | 125328375 | Intron 1 | | 6 | T, 0.1772 | CC/CT/TT | 511/221/23 | 136.58/137.05/136.75 | 27.68/28.31/22.08 | 0.1215 | 0.6158 | 0.8437 | 0.964 |
| p22116 | rs12370382 | 125327404 | Intron 1 | | 1f | A, 0.0645 | GG/GA/AA | 650/89/3 | 135.9/139.94/107.7 | 27.29/31.23/14.64 | 0.5264 | 0.9717 | 0.5882 | 0.859 |
| p22168 | rs7137797 | 125327352 | Intron 1 | | 2b | C, 0.3977 | CC/TC/TT | 112/358/262 | 139.7/135.97/135.95 | 27.57/26.56/28.95 | 0.5066 | 0.4888 | 0.3003 | 0.776 |
| p22331 | rs6488944 | 125327189 | Intron 1 | | 4 | G, 0.1474 | TT/TG/GG | 524/191/12 | 136.47/138.31/134.29 | 27.51/28.77/27.05 | 0.3684 | 0.6902 | 0.5936 | 0.859 |
| p22675 | rs12425134 | 125326845 | Intron 1 | | 4 | T, 0.0526 | TT/GT/GG | 2/74/668 | 149.31/139.33/136.52 | 0.28/23.43/28.01 | 1.1151 | 1.0532 | 0.2900 | 0.776 |
| p28137 | rs12229555 | 125321383 | Intron 1 | | 7 | G, 0.3896 | GG/AG/AA | 100/373/265 | 134.51/136.62/138.27 | 24.37/27.67/28.92 | -0.5752 | 0.5022 | 0.2524 | 0.776 |
| p28692 | rs4765622 | 125320828 | Intron 1 | | 5 | T, 0.2565 | TT/CT/CC | 42/298/406 | 134.55/140.06/135.18 | 28.51/29.32/26.22 | 0.7871 | 0.5534 | 0.1553 | 0.642 |
| p28957 | rs11057844 | 125320563 | Intron 1 | | 5 | A, 0.2362 | GG/GA/AA | 433/269/40 | 136.15/136.91/142.06 | 28.60/26.35/29.06 | 0.6107 | 0.5626 | 0.2781 | 0.776 |
| p29749 | rs10846751 | 125319771 | Intron 1 | | 7 | T, 0.4492 | TT/CT/CC | 142/366/215 | 132.92/137.38/138.1 | 25.65/27.79/28.67 | -0.7763 | 0.4856 | 0.1104 | 0.575 |
| p31072 | rs10846749 | 125318448 | Intron 1 | | 4 | G, 0.4461 | CC/CG/GG | 226/371/146 | 139.23/136.07/134.5 | 28.54/28.23/26.18 | -0.8005 | 0.4818 | 0.0970 | 0.575 |
| p31938 | rs10744182 | 125317582 | Intron 1 | | 5 | A, 0.1837 | AA/GA/GG | 25/226/493 | 131.55/136.6/137.11 | 27.78/27.47/28.13 | -0.4389 | 0.6139 | 0.4748 | 0.826 |
| p32129 | rs10773107 | 125317391 | Intron 1 | | 7 | T, 0.1009 | TT/GT/GG | 9/131/605 | 151.68/137.5/136.38 | 25.54/28.08/27.45 | 0.8714 | 0.7738 | 0.2605 | 0.776 |
| p32273 | rs12580803 | 125317247 | Intron 1 | | 5 | C, 0.1006 | TT/TC/CC | 627/117/18 | 135.83/140.74/140.03 | 27.84/27.87/24.49 | 1.2550 | 0.7258 | 0.0842 | 0.558 |
| p32290 | rs10744181 | 125317230 | Intron 1 | | 5 | C, 0.1238 | TT/TC/CC | 551/143/18 | 136.61/139.47/129.44 | 27.77/28.30/23.29 | 0.1584 | 0.7002 | 0.8211 | 0.954 |
| p32395 | rs12581963 | 125317125 | Intron 1 | | 5 | T, 0.1314 | TT/CT/CC | 11/177/570 | 134.74/134.94/137.17 | 27.03/27.17/27.89 | -0.6483 | 0.7013 | 0.3556 | 0.776 |
| p32750 | rs7967521 | 125316770 | Intron 1 | | 7 | G, 0.3425 | GG/AG/AA | 91/304/315 | 135.77/138.19/136.64 | 27.06/27.89/28.28 | 0.0564 | 0.5011 | 0.9104 | 0.975 |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p32777 | rs11057841 | 125316743 | Intron 1 | | 7 | A, 0.2805 | GG/GA/AA | 374/303/52 | 136.83/137.07/136.83 | 27.62/27.46/29.55 | 0.0373 | 0.5407 | 0.9450 | 0.992 |
| p32860 | rs7967406 | 125316660 | Intron 1 | | 6 | C, 0.0991 | CC/AC/AA | 9/135/616 | 142.53/137.42/136.54 | 29.57/26.48/28.02 | 0.4768 | 0.7717 | 0.5369 | 0.849 |
| p33531 | rs11057838 | 125315989 | Intron 1 | | 7 | A, 0.2278 | CC/CA/AA | 440/259/40 | 136.66/137.21/137.74 | 28.28/26.63/27.18 | 0.1993 | 0.5594 | 0.7218 | 0.914 |
| p36094 | rs11608336 | 125313426 | Intron 1 | | 4 | A, 0.1543 | GG/GA/AA | 526/195/16 | 136.58/137.43/130.93 | 26.64/30.54/19.66 | -0.0956 | 0.6599 | 0.8848 | 0.965 |
| p36461 | rs4765178 | 125313059 | Intron 1 | | 4 | T, 0.1671 | TT/CT/CC | 18/205/497 | 143.57/137.38/136.71 | 28.60/27.50/28.03 | 0.5087 | 0.6538 | 0.4368 | 0.811 |
| p36908 | rs10846745 | 125312612 | Intron 1 | | 4 | G, 0.3257 | GG/CG/CC | 78/335/337 | 133.81/138.5/136.23 | 26.52/28.07/27.17 | 0.0089 | 0.5014 | 0.9859 | 0.997 |
| p37095 | rs10846744 | 125312425 | Intron 1 | | 4 | G, 0.3056 | CC/CG/GG | 350/350/56 | 137.16/135.63/141.68 | 27.54/27.45/30.05 | 0.1538 | 0.5335 | 0.7732 | 0.914 |
| p41632 | rs6488943 | 125307888 | Intron 1 | | 5 | C, 0.2954 | CC/AC/AA | 52/312/340 | 139.91/137.51/137.18 | 25.93/27.34/27.81 | 0.3025 | 0.5440 | 0.5783 | 0.857 |
| p42467 | rs11057830 | 125307053 | Intron 1 | | 7 | T, 0.1523 | TT/CT/CC | 14/200/533 | 137.96/133.93/137.64 | 41.61/27.21/27.42 | -0.9351 | 0.6668 | 0.1612 | 0.642 |
| p45516 | rs1902569 | 125304004 | Intron 1 | | 5 | A, 0.1544 | AA/GA/GG | 18/191/523 | 142.37/137.21/136.5 | 23.01/25.54/28.50 | 0.5127 | 0.6537 | 0.4331 | 0.811 |
| p48969 | rs2343394 | 125300551 | Intron 2 | | 5 | T, 0.1898 | TT/CT/CC | 32/226/501 | 138.49/138.05/135.89 | 20.81/26.09/28.67 | 0.6411 | 0.5825 | 0.2714 | 0.776 |
| p49537 | rs7305310 | 125299983 | Intron 2 | | 5 | T, 0.1007 | CC/CT/TT | 601/118/16 | 136.94/137.31/131.4 | 28.45/24.39/24.26 | -0.2265 | 0.7408 | 0.7599 | 0.914 |
| p49570 delC | rs145376237 | 125299950 | Intron 2 | | 5 | delC, 0.2276 | DD/WD/WW | 37/260/437 | 132.41/139.59/135.62 | 31.14/26.82/28.01 | 0.4929 | 0.5706 | 0.3880 | 0.776 |
| p49690 | rs4765615 | 125299830 | Intron 2 | | 5 | A, 0.4426 | AA/GA/GG | 157/323/244 | 132.82/137.57/138.51 | 31.67/26.38/26.87 | -0.9139 | 0.4614 | **0.0480** | 0.502 |
| p50151 | rs2278986 | 125299369 | Intron 3 | | 5 | C, 0.1933 | CC/TC/TT | 33/226/494 | 142.72/137.51/135.83 | 21.16/25.91/28.90 | 0.8568 | 0.5828 | 0.1419 | 0.631 |
| p51888 | rs7138304 | 125297632 | Intron 4 | | 2b | T, 0.1079 | TT/CT/CC | 16/130/593 | 130.58/138.07/136.64 | 24.75/23.20/28.33 | -0.0031 | 0.7159 | 0.9965 | 0.997 |
| p52096 | rs10846739 | 125297424 | Intron 4 | | 3a | G, 0.4693 | GG/AG/AA | 172/342/221 | 135.77/137.19/136.77 | 28.27/27.28/27.46 | -0.1537 | 0.4586 | 0.7377 | 0.914 |
| p52556 | rs11057820 | 125296964 | Intron 4 | | 5 | A, 0.1000 | AA/GA/GG | 12/126/619 | 127.43/134.64/137.37 | 29.15/29.91/27.33 | -1.1588 | 0.7609 | 0.1282 | 0.623 |
| p52610 | rs10846738 | 125296910 | Intron 4 | | 4 | T, 0.1349 | TT/CT/CC | 13/172/553 | 130.76/135.7/137.19 | 33.53/27.12/27.43 | -0.6490 | 0.6886 | 0.3462 | 0.776 |
| p52956 | rs77740046 | 125296564 | Intron 4 | | 5 | T, 0.0546 | CC/CT/TT | 657/75/3 | 136.98/135.38/134.32 | 28.41/22.14/29.35 | -0.4428 | 1.0350 | 0.6689 | 0.905 |
| p53359 | rs112371713 | 125296161 | Intron 5 | | 5 | A, 0.1243 | AA/GA/GG | 10/161/554 | 129.69/140.18/136.1 | 21.21/27.65/28.05 | 0.8266 | 0.7340 | 0.2605 | 0.776 |
| p53790 | rs4765614 | 125295730 | Intron 5 | | 5 | A, 0.2653 | GG/GA/AA | 402/277/59 | 137.25/136.67/133.06 | 28.76/25.80/28.38 | -0.4621 | 0.5232 | 0.3775 | 0.776 |
| p54492 | rs61762481 | 125295028 | Intron 5 | | 4 | A, 0.1005 | AA/GA/GG | 8/137/618 | 148.92/134.86/137.03 | 32.80/27.06/27.71 | -0.1546 | 0.7735 | 0.8416 | 0.964 |
| p55923 | rs838900 | 125293597 | Intron 6 | | 7 | A, 0.3921 | AA/GA/GG | 115/346/282 | 138.57/136.64/135.92 | 31.53/27.67/26.43 | 0.3606 | 0.4823 | 0.4549 | 0.823 |
| p55963 | rs7134858 | 125293557 | Intron 6 | | 6 | T, 0.1560 | TT/CT/CC | 24/190/537 | 147.89/139.6/135.05 | 30.36/25.98/28.12 | 1.7537 | 0.6260 | **0.0052** | 0.292 |
| p56845 | rs838902 | 125292675 | Intron 6 | | 5 | G, 0.4249 | AA/AG/GG | 256/350/143 | 136.96/138.11/134.33 | 28.98/27.23/26.41 | -0.3052 | 0.4662 | 0.5129 | 0.840 |
| p57107 | rs5892 | 125292413 | Exon 7 | Phe301Phe | 4 | T, 0.0589 | CC/CT/TT | 663/84/1 | 136.93/135.46/135.43 | 27.81/27.27/NA | -0.4697 | 1.0308 | 0.6488 | 0.900 |
| p57508 | rs71458866 | 125292012 | Intron 7 | | 4 | A, 0.1130 | AA/GA/GG | 12/147/602 | 136.02/134.65/137.08 | 32.39/27.43/27.74 | -0.6325 | 0.7294 | 0.3861 | 0.776 |
| p57592 | rs838903 | 125291928 | Intron 7 | | 4 | A, 0.3763 | AA/GA/GG | 113/336/297 | 132.17/137.53/138 | 26.07/27.56/28.72 | -0.7661 | 0.4801 | 0.1109 | 0.575 |
| p58514 | rs838905 | 125291006 | Intron 7 | | 4 | C, 0.4329 | CC/TC/TT | 149/352/250 | 134.23/138.05/137.42 | 26.43/27.51/28.81 | -0.4213 | 0.4644 | 0.3646 | 0.776 |
| p58664 | rs865716 | 125290856 | Intron 7 | | 5 | T, 0.2708 | AA/AT/TT | 395/272/63 | 136.68/135.18/144.1 | 28.06/26.58/28.78 | 0.5369 | 0.5185 | 0.3008 | 0.776 |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p60255 | rs3782287 | 125289265 | Intron 7 | | 5 | T, 0.2831 | CC/CT/TT | 384/314/55 | 136.2/136.74/139.92 | 26.41/28.49/33.12 | 0.3715 | 0.5324 | 0.4856 | 0.826 |
| p61872 | rs838909 | 125287648 | Intron 7 | | 4 | T, 0.2199 | CC/CT/TT | 453/251/36 | 135.22/138.63/139.38 | 27.18/28.46/28.12 | 0.9232 | 0.5699 | 0.1056 | 0.575 |
| p62140 | rs838910 | 125287380 | Intron 7 | | 5 | T, 0.3047 | GG/GT/TT | 358/304/72 | 135.54/139.34/130.23 | 27.71/28.09/24.10 | -0.0755 | 0.5087 | 0.8821 | 0.965 |
| p62409 | rs838911 | 125287111 | Intron 7 | | 5 | T, 0.4211 | CC/CT/TT | 251/346/135 | 137.46/138/132.71 | 28.39/28.04/25.39 | -0.6245 | 0.4748 | 0.1888 | 0.694 |
| p62615 | rs7138386 | 125286905 | Intron 7 | | 5 | C, 0.1137 | TT/TC/CC | 571/141/11 | 137.41/132.86/147.97 | 27.69/26.95/32.74 | -0.6495 | 0.7473 | 0.3851 | 0.776 |
| p63483 | rs838912 | 125286037 | Intron 7 | | 7 | A, 0.0867 | AA/GA/GG | 6/119/624 | 147.92/141.68/136.06 | 22.36/28.42/27.33 | 1.8700 | 0.8230 | **0.0234** | 0.397 |
| p64772 | rs5888 | 125284748 | Exon 8 | Ala350Ala | 3a | T, 0.0961 | CC/CT/TT | 614/130/8 | 135.6/141.63/150.76 | 27.57/28.87/21.81 | 2.0962 | 0.7888 | **0.0080** | 0.292 |
| p64923 | rs838915 | 125284597 | Intron 8 | | 5 | A, 0.1435 | AA/CA/CC | 19/179/547 | 128.24/137.41/136.97 | 24.55/26.33/28.38 | -0.3684 | 0.6595 | 0.5766 | 0.857 |
| p65999 | rs12819677 | 125283521 | Intron 8 | | 6 | A, 0.2813 | GG/GA/AA | 375/309/52 | 135.65/137.67/140.14 | 26.43/28.07/31.39 | 0.6769 | 0.5339 | 0.2052 | 0.716 |
| p67439 | rs961170 | 125282081 | Intron 8 | | 4 | A, 0.0893 | AA/GA/GG | 11/108/615 | 127.38/135.18/137.19 | 30.33/28.13/27.62 | -0.9808 | 0.8009 | 0.2211 | 0.743 |
| p67700 | rs1726374 | 125281820 | Intron 8 | | 7 | A, 0.1933 | GG/GA/AA | 500/226/32 | 136.28/137.42/137.84 | 27.77/28.32/24.61 | 0.3249 | 0.5867 | 0.5798 | 0.857 |
| p69013 | rs7135117 | 125280507 | Intron 8 | | 7 | G, 0.2901 | GG/AG/AA | 74/275/383 | 139.15/137.26/135.9 | 27.28/28.32/27.33 | 0.5010 | 0.5031 | 0.3196 | 0.776 |
| p69699 | rs10396210 | 125279821 | Intron 8-splice site | | 4 | A, 0.1511 | AA/GA/GG | 17/188/527 | 133.13/133.32/138.01 | 24.50/25.99/28.45 | -1.2921 | 0.6639 | 0.0520 | 0.505 |
| p69995 delC | rs5801571 | 125279525 | Intron 9 | | 5 | delC, 0.2761 | DD/WD/WW | 63/282/394 | 138.36/136.65/136.08 | 29.51/27.84/27.69 | 0.2920 | 0.5230 | 0.5768 | 0.857 |
| p71867 | rs7954022 | 125277653 | Intron 9 | | 5 | T, 0.1323 | TT/CT/CC | 12/172/556 | 138.36/139.03/136.02 | 28.42/27.41/27.66 | 0.8502 | 0.6988 | 0.2241 | 0.743 |
| p72197 | rs838861 | 125277323 | Intron 9 | | 7 | G, 0.3777 | AA/AG/GG | 304/310/122 | 138.19/135.01/138.6 | 28.51/26.54/28.32 | -0.1507 | 0.4657 | 0.7464 | 0.914 |
| p72777 | rs838862 | 125276743 | Intron 9 | | 5 | T, 0.0887 | CC/CT/TT | 611/116/7 | 136.49/140.05/130.13 | 27.50/27.25/33.12 | 0.7012 | 0.8218 | 0.3938 | 0.776 |
| p75766 | rs838866 | 125273754 | Intron 9 | | 6 | C, 0.2116 | TT/TC/CC | 466/240/37 | 136.81/137.6/134.32 | 27.72/27.49/29.38 | -0.0497 | 0.5699 | 0.9306 | 0.989 |
| p75778 | rs7301120 | 125273742 | Intron 9 | | 6 | T, 0.1135 | TT/CT/CC | 9/146/569 | 153.8/135.98/136.84 | 34.57/28.15/27.34 | 0.3767 | 0.7539 | 0.6174 | 0.884 |
| p76757 | rs9919713 | 125272763 | Intron 9 | | 6 | T, 0.4390 | AA/AT/TT | 243/353/148 | 137.25/137.33/135.86 | 29.00/27.02/27.54 | -0.1860 | 0.4696 | 0.6921 | 0.905 |
| p77251 | rs34339961 | 125272269 | Intron 9 | | 6 | T, 0.1177 | AA/AT/TT | 573/150/11 | 136.68/136.84/153.06 | 27.85/28.26/33.32 | 0.7120 | 0.7432 | 0.3383 | 0.776 |
| p77842 | rs2272310 | 125271678 | Intron 10 | | 5 | A, 0.0807 | AA/GA/GG | 5/110/642 | 136.12/139.14/136.1 | 4.96/29.15/27.59 | 0.8339 | 0.8661 | 0.3359 | 0.776 |
| p78402 | rs838898 | 125271118 | Intron 10 | | 5 | A, 0.0714 | AA/GA/GG | 7/86/601 | 124.15/135.76/137.49 | 33.52/24.94/28.40 | -0.9806 | 0.9240 | 0.2889 | 0.776 |
| p78430 | rs838897 | 125271090 | Intron 10 | | 5 | G, 0.3830 | GG/CG/CC | 121/315/299 | 135.15/137.21/136.75 | 28.01/28.01/27.48 | -0.1887 | 0.4709 | 0.6887 | 0.905 |
| p78747 | rs2293440 | 125270773 | Intron 11 | | 5 | C, 0.4112 | CC/TC/TT | 126/348/258 | 136.06/136.3/137.79 | 27.37/26.42/29.64 | -0.2984 | 0.4810 | 0.5352 | 0.849 |
| p79721 | rs838896 | 125269799 | Intron 11 | | 5 | C, 0.3104 | GG/GC/CC | 357/324/71 | 135.09/137.26/143.1 | 28.43/26.98/24.61 | 1.1147 | 0.5056 | **0.0278** | 0.420 |
| p79828 | rs838895 | 125269692 | Intron 11 | | 5 | G, 0.3171 | GG/CG/CC | 73/325/345 | 142.03/137.79/134.49 | 25.40/26.62/28.83 | 1.2206 | 0.5074 | **0.0164** | 0.319 |
| p80045 | rs838893 | 125269475 | Intron 11 | | 5 | A, 0.3244 | GG/GA/AA | 344/328/80 | 135.11/137.56/140.43 | 29.04/26.78/25.93 | 0.8859 | 0.5009 | 0.0774 | 0.554 |
| p83547 | rs838887 | 125265973 | Intron 12 | | 5 | G, 0.4564 | CC/CG/GG | 227/358/157 | 136.15/137.97/135.3 | 25.51/27.56/30.61 | -0.1113 | 0.4676 | 0.8119 | 0.952 |
| p83884 | rs701106 | 125265636 | Intron 12 | | 5 | T, 0.2597 | TT/CT/CC | 51/291/412 | 142.85/138.67/134.71 | 28.78/29.50/26.27 | 1.2967 | 0.5352 | **0.0156** | 0.319 |
| p86276 | rs747155 | 125263244 | Intron 12 | | 2b | T, 0.1495 | TT/CT/CC | 17/191/541 | 131.8/136.97/136.81 | 35.42/26.39/28.03 | -0.2164 | 0.6605 | 0.7433 | 0.914 |

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|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p86481 | rs701103 | 125263039 | Exon 13-3' UTR | Gly499Arg (isoform 2) | 5 | A, 0.2451 | AA/GA/GG | 50/265/428 | 132.37/138.77/136.53 | 27.64/27.23/28.05 | -0.0074 | 0.5400 | 0.9891 | 0.997 |
| p87011 | rs58032386 | 125262509 | Exon 13-3' UTR | | 2a | T, 0.1417 | CC/CT/TT | 551/186/14 | 137.07/137/129.91 | 28.90/24.33/24.12 | -0.2769 | 0.6776 | 0.6829 | 0.905 |
| p87723 | rs838881 | 125261797 | 3' flanking | | 6 | T, 0.3183 | TT/CT/CC | 65/344/338 | 134.34/137.04/136.76 | 28.25/27.36/28.31 | -0.1897 | 0.5263 | 0.7187 | 0.914 |
| p87749 | rs76465225 | 125261771 | 3' flanking | | 7 | A, 0.0844 | AA/GA/GG | 7/111/629 | 128.6/137.32/136.7 | 23.34/27.81/27.96 | -0.1205 | 0.8434 | 0.8865 | 0.965 |
| p87927 | rs838880 | 125261593 | 3' flanking | | 5 | A, 0.2414 | AA/GA/GG | 40/280/423 | 134.92/136.85/137.12 | 29.87/27.16/28.26 | -0.2130 | 0.5636 | 0.7056 | 0.914 |
| **MAF between 1-5%** | | | | | | | | | | | | | | |
| p1316 | rs10396208 | 125348204 | Exon 1 | Cys21Cys | 2b | T, 0.0476 | CC/CT/TT | 657/60/4 | 136.18/145.72/126.27 | 27.33/32.28/23.99 | 2.0253 | 1.0962 | 0.0651 | 0.521 |
| p1419 | rs201717369 | 125348101 | Intron 1 | | 4 | A, 0.0121 | GA/GG | 15/713 | 143.53/136.72 | 33.66/27.75 | 2.1508 | 2.3880 | 0.3681 | 0.776 |
| p7650 | rs11615630 | 125341870 | Intron 1 | | 5 | A, 0.0436 | GG/GA | 695/68 | 137.43/130.27 | 27.44/30.31 | -2.4339 | 1.1578 | **0.0359** | 0.436 |
| p45627 | rs12297372 | 125303893 | Intron 1 | | 5 | G, 0.0487 | GG/AG/AA | 1/71/661 | 132.07/139.72/136.41 | NA/29.73/27.51 | 0.9720 | 1.1101 | 0.3816 | 0.776 |
| p46964 | rs114061302 | 125302556 | Intron 1 | | 4 | A, 0.0388 | AA/GA/GG | 1/58/696 | 92.79/139.86/136.49 | NA/31.62/27.47 | 0.4839 | 1.2105 | 0.6895 | 0.905 |
| p50118 | rs58710319 | 125299402 | Intron 3 | | 5 | T, 0.0208 | CT/CC | 32/719 | 146.19/136.45 | 32.51/27.51 | 3.1376 | 1.6463 | 0.0571 | 0.517 |
| p50380 | rs141748317 | 125299140 | Intron 3 | | 2b | G, 0.0112 | AA/AG | 727/17 | 136.64/137.87 | 27.67/30.07 | 0.3920 | 2.2372 | 0.8609 | 0.965 |
| p50489 | rs61320152 | 125299031 | Intron 3 | | 4 | T, 0.0257 | GG/GT | 704/39 | 136.63/143.42 | 27.39/31.34 | 2.1813 | 1.4908 | 0.1439 | 0.631 |
| p54445 | rs60910935 | 125295075 | Intron 5 | | 4 | G, 0.0418 | AA/AG/GG | 656/57/2 | 137.28/132.79/148.43 | 28.47/22.46/21.21 | -0.9880 | 1.1918 | 0.4074 | 0.791 |
| p54475 | rs60227139 | 125295045 | Intron 5 | | 4 | T, 0.0437 | CC/CT/TT | 676/61/2 | 137.11/133.52/148.4 | 28.23/22.68/21.21 | -0.7550 | 1.1492 | 0.5114 | 0.840 |
| p77682 | rs150082885 | 125271838 | Intron 10 | | 5 | G, 0.0106 | AA/AG/GG | 721/14/1 | 137.13/122.98/156.01 | 27.77/29.30/NA | -2.9877 | 2.1744 | 0.1698 | 0.642 |
| p78791 | rs75289200 | 125270729 | Intron 11 | | 5 | C, 0.0321 | TC/TT | 46/685 | 147.19/136.06 | 27.99/27.68 | 3.6568 | 1.3876 | **0.0086** | 0.292 |
| p82019 | rs838890 | 125267501 | Intron 11 | | 5 | T, 0.0320 | CC/CT/TT | 690/41/2 | 137.12/135.58/112.02 | 27.74/28.19/72.83 | -1.1864 | 1.3456 | 0.3782 | 0.776 |
| p82340 | rs77483223 | 125267180 | Intron 12 | | 5 | A, 0.0231 | GA/GG | 35/705 | 132.2/137.01 | 25.03/27.96 | -1.5601 | 1.5901 | 0.3268 | 0.776 |
| p82434 | rs838889 | 125267086 | Intron 12 | | 5 | C, 0.0315 | CC/TC/TT | 2/41/702 | 112.08/136.37/136.99 | 72.83/26.99/27.63 | -0.9096 | 1.3374 | 0.4967 | 0.834 |
| p86245 | rs188375019 | 125263275 | Intron 12 | | 4 | T, 0.0341 | CC/CT | 696/50 | 136.54/142.03 | 27.69/26.72 | 1.8399 | 1.3314 | 0.1674 | 0.642 |
| p86316 | rs701104 | 125263204 | Intron 12 | | 4 | T, 0.0487 | TT/GT/GG | 2/64/658 | 128.07/135.86/137.43 | 50.49/29.05/27.70 | -0.6627 | 1.1303 | 0.5579 | 0.857 |
| p86967 | rs187492239 | 125262553 | Exon 13-3' UTR | | 4 | G, 0.0355 | AA/AG | 692/52 | 136.5/140.59 | 27.82/26.49 | 1.3818 | 1.3114 | 0.2924 | 0.776 |
| p87611 | rs190688220 | 125261909 | 3' flanking | | 4 | T, 0.0316 | CC/CT | 697/46 | 136.41/141.77 | 27.77/27.47 | 1.7851 | 1.3894 | 0.1993 | 0.713 |
| p87681 | rs838883 | 125261839 | 3' flanking | | 5 | A, 0.0459 | AA/GA/GG | 1/62/659 | 163.53/136.09/137.17 | NA/28.31/27.91 | -0.0535 | 1.1812 | 0.9639 | 0.993 |
| **MAF ≤1%** | | | | | | | | | | | | | | |
| p1048 insC (1048_1049) | | 125348472 | Exon 1-5' UTR | | 2a | insC, 0.0079 | WI/WW | 12/729 | 134.79/136.97 | 31.94/27.67 | -0.7733 | 2.6557 | 0.7710 | 0.914 |
| p49759 | rs146272788 | 125299761 | Intron 2 | | 5 | T, 0.0020 | CC/CT | 731/3 | 137.03/141.39 | 27.75/17.56 | 1.5883 | 5.2643 | 0.7630 | 0.914 |
| p49978 | rs5891 | 125299542 | Exon 3 | Val135Ile | 5 | A, 0.0058 | GA/GG | 9/754 | 154.31/136.52 | 39.14/27.50 | 5.6762 | 3.0464 | 0.0628 | 0.521 |

| | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| p50024 | rs368880622 | 125299496 | Intron 3 | | 5 | T, 0.0026 | GG/GT | 742/4 | 136.83/142.91 | 27.51/57.60 | 1.6012 | 4.5586 | 0.7255 | 0.914 |
| p50954 | | 125298566 | Intron 4 | | 5 | C, 0.0007 | TC/TT | 1/742 | 110.14/136.79 | NA/27.75 | -8.7626 | 9.1297 | 0.3375 | 0.776 |
| p52919 | | 125296601 | Intron 4 | | 5 | T, 0.0013 | GG/GT | 741/2 | 136.81/97.42 | 27.74/18.38 | -13.4137 | 6.4689 | **0.0385** | 0.436 |
| p52995 | rs113910315 | 125296525 | Intron 4-splice site | | 5 | G, 0.0020 | TG/TT | 3/746 | 138.8/136.77 | 34.93/27.70 | 0.6563 | 5.2739 | 0.9010 | 0.973 |
| p53372 | rs115604379 | 125296148 | Intron 5 | | 5 | T, 0.0066 | CC/CT | 735/10 | 136.57/149.32 | 27.83/17.13 | 4.3130 | 2.9092 | 0.1386 | 0.631 |
| p54611 | | 125294909 | Intron 5 | | 4 | C, 0.0007 | TC/TT | 1/748 | 80.62/136.83 | NA/27.66 | -19.2831 | 9.0970 | **0.0344** | 0.436 |
| p54627 | | 125294893 | Intron 5 | | 4 | C, 0.0020 | GC/GG | 3/744 | 147.45/136.54 | 23.34/27.76 | 3.6910 | 5.2836 | 0.4850 | 0.826 |
| p54856 | | 125294664 | Intron 6 | | 4 | T, 0.0007 | CC/CT | 748/1 | 136.87/67.98 | 27.61/NA | -24.0757 | 9.0781 | **0.0082** | 0.292 |
| p57004 | rs187562853 | 125292516 | Intron 6 | | 4 | A, 0.0098 | GG/GA | 731/15 | 136.33/146.22 | 27.82/26.41 | 3.2853 | 2.3865 | 0.1690 | 0.642 |
| p77181 | rs146246031 | 125272339 | Intron 9 | | 7 | C, 0.0053 | TC/TT | 8/734 | 138.87/137.05 | 40.35/27.46 | 0.4564 | 3.2210 | 0.8874 | 0.965 |
| p77381 | rs138499966 | 125272139 | Intron 9 | | 6 | C, 0.0046 | TC/TT | 7/741 | 135.79/136.78 | 22.32/27.79 | -0.2236 | 3.4610 | 0.9485 | 0.992 |
| p77620 | rs377124254 | 125271900 | Intron 10 | | 5 | A, 0.0007 | GA/GG | 1/741 | 181.07/136.69 | NA/27.81 | 14.4685 | 9.1465 | 0.1141 | 0.575 |
| p77704 | | 125271816 | Intron 10 | | 5 | A, 0.0040 | CA/CC | 6/731 | 127.91/136.87 | 31.82/27.82 | -3.0744 | 3.7716 | 0.4152 | 0.795 |
| p78255 | rs184052375 | 125271265 | Intron 10 | | 4 | G, 0.0072 | AA/AG | 740/9 | 136.63/147.01 | 27.63/34.72 | 3.3097 | 3.0552 | 0.2790 | 0.776 |
| p81863 | rs185445624 | 125267657 | Intron 11 | | 5 | A, 0.0020 | GA/GG | 3/745 | 129.03/136.84 | 26.05/27.73 | -2.5166 | 5.2725 | 0.6333 | 0.897 |
| p82264 | rs141545424 | 125267256 | Exon 12 | Gly501Gly | 5 | A, 0.0007 | CA/CC | 1/745 | 181.22/136.74 | NA/27.71 | 14.4986 | 9.1095 | 0.1119 | 0.575 |
| p82369 | rs75446635 | 125267151 | Intron 12 | | 5 | A, 0.0059 | GA/GG | 9/739 | 144.2/136.77 | 41.63/27.47 | 2.2239 | 3.0489 | 0.4660 | 0.823 |
| p87266 | rs150512235 | 125262254 | Exon 13-3' UTR[f] | | 4 | C, 0.0057 | TC/TT | 9/756 | 137.13/136.56 | 38.11/27.67 | 0.0318 | 3.0667 | 0.9917 | 0.997 |
| p87694 | | 125261826 | 3' flanking | | 5 | T, 0.0020 | CC/CT | 731/3 | 136.86/145.93 | 27.92/24.62 | 3.0751 | 5.3006 | 0.5620 | 0.857 |

del/D, deletion; FDR, false discovery rate; ins/I, insertion; MA, minor allele; MAF, minor allele frequency; NA, not analyzed; RegDB, RegulomeDB score; SD, standard deviation; SE, standard error; SNP, single nucleotide polymorphism; UTR, untranslated region; W, wild type allele on RefSeq for insertion and deletion variations.

All alleles on reverse strand. Splice site is defined as ± 20 bp from the start or end of an exon.

ApoA-I values were Box-Cox transformed. Results were adjusted for covariates: sex and age.

Nominally significant *P*-values (*P* <0.05) and FDR values that passed the threshold (FDR <0.20) are shown in **bold**.

[a, c] Based on the RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics).

[b] dbSNP build 139: GRCh37.p10; 10 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_AB).

[d] Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table B4 or can be seen at http://regulome.stanford.edu/help.

[e] FDR is referred to a *q*-value from Benjamini-Hochberg procedure.

[f] Close to the miRNA-145 seed binding site based on TargetScanHuman (version 6.2, http://www.targetscan.org/).

## Table B11. Haplotype association results of 136 *SCARB1* genotyped variants (haplotype windows and global *P*-values) for HDL-C and ApoA-I.

| | | | | | | HDL-C | | | | ApoA-I | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| wind | wind SNP | SNP Name[a]-SNP ID[b] /Chr12 Position[c] | Location | MA, MAF | Genotype | Genotype Count | β | Single-site *P* | Global *P* | Genotype Count | β | Single-site *P* | Global *P* |
| 1 | SNP 1 | p1048insC-chr12_125348472 | Exon 1-5' UTR | insC, 0.0079 | WI/WW | 12/723 | -0.9389 | 0.3785 | 0.3506 | 12/729 | -0.7733 | 0.7710 | 0.1160 |
| 1 | SNP 2 | p1265-rs2070242 | Exon 1 | T, 0.1284 | CC/CT/TT | 541/163/10 | -0.5040 | 0.0833 | | 545/165/10 | -1.2535 | 0.0862 | |
| 1 | SNP 3 | p1316-rs10396208 | Exon 1 | T, 0.0476 | CC/CT/TT | 652/58/4 | 0.2987 | 0.5001 | | 657/60/4 | 2.0253 | 0.0651 | |
| 1 | SNP 4 | p1419-rs201717369 | Intron 1 | A, 0.0121 | AA/GA/GG | 1/14/707 | 0.4892 | 0.5748 | | 0/15/713 | 2.1508 | 0.3681 | |
| 2 | SNP 1 | p1265-rs2070242 | Exon 1 | T, 0.1284 | CC/CT/TT | 541/163/10 | -0.5040 | 0.0833 | 0.6456 | 545/165/10 | -1.2535 | 0.0862 | 0.1454 |
| 2 | SNP 2 | p1316-rs10396208 | Exon 1 | T, 0.0476 | CC/CT/TT | 652/58/4 | 0.2987 | 0.5001 | | 657/60/4 | 2.0253 | 0.0651 | |
| 2 | SNP 3 | p1419-rs201717369 | Intron 1 | A, 0.0121 | AA/GA/GG | 1/14/707 | 0.4892 | 0.5748 | | 0/15/713 | 2.1508 | 0.3681 | |
| 2 | SNP 4 | p4012-rs7139401 | Intron 1 | C, 0.4386 | TT/TC/CC | 232/343/147 | 0.0877 | 0.6453 | | 236/350/148 | -0.0802 | 0.8654 | |
| 3 | SNP 1 | p1316-rs10396208 | Exon 1 | T, 0.0476 | CC/CT/TT | 652/58/4 | 0.2987 | 0.5001 | 0.8268 | 657/60/4 | 2.0253 | 0.0651 | 0.2992 |
| 3 | SNP 2 | p1419-rs201717369 | Intron 1 | A, 0.0121 | AA/GA/GG | 1/14/707 | 0.4892 | 0.5748 | | 0/15/713 | 2.1508 | 0.3681 | |
| 3 | SNP 3 | p4012-rs7139401 | Intron 1 | C, 0.4386 | TT/TC/CC | 232/343/147 | 0.0877 | 0.6453 | | 236/350/148 | -0.0802 | 0.8654 | |
| 3 | SNP 4 | p5055-rs11057869 | Intron 1 | A, 0.0740 | AA/GA/GG | 7/96/638 | 0.0976 | 0.7828 | | 7/95/650 | 0.5638 | 0.5247 | |
| 4 | SNP 1 | p1419-rs201717369 | Intron 1 | A, 0.0121 | AA/GA/GG | 1/14/707 | 0.4892 | 0.5748 | 0.6474 | 0/15/713 | 2.1508 | 0.3681 | 0.7873 |
| 4 | SNP 2 | p4012-rs7139401 | Intron 1 | C, 0.4386 | TT/TC/CC | 232/343/147 | 0.0877 | 0.6453 | | 236/350/148 | -0.0802 | 0.8654 | |
| 4 | SNP 3 | p5055-rs11057869 | Intron 1 | A, 0.0740 | AA/GA/GG | 7/96/638 | 0.0976 | 0.7828 | | 7/95/650 | 0.5638 | 0.5247 | |
| 4 | SNP 4 | p6600-rs12831105 | Intron 1 | T, 0.1188 | CC/CT/TT | 566/151/11 | 0.3570 | 0.2239 | | 571/152/10 | -0.5681 | 0.4412 | |
| 5 | SNP 1 | p4012-rs7139401 | Intron 1 | C, 0.4386 | TT/TC/CC | 232/343/147 | 0.0877 | 0.6453 | 0.6759 | 236/350/148 | -0.0802 | 0.8654 | 0.2798 |
| 5 | SNP 2 | p5055-rs11057869 | Intron 1 | A, 0.0740 | AA/GA/GG | 7/96/638 | 0.0976 | 0.7828 | | 7/95/650 | 0.5638 | 0.5247 | |
| 5 | SNP 3 | p6600-rs12831105 | Intron 1 | T, 0.1188 | CC/CT/TT | 566/151/11 | 0.3570 | 0.2239 | | 571/152/10 | -0.5681 | 0.4412 | |
| 5 | SNP 4 | p7650-rs11615630 | Intron 1 | A, 0.0436 | GG/GA | 685/67 | 0.3382 | 0.4702 | | 695/68 | -2.4339 | 0.0359 | |
| 6 | SNP 1 | p5055-rs11057869 | Intron 1 | A, 0.0740 | AA/GA/GG | 7/96/638 | 0.0976 | 0.7828 | 0.7353 | 7/95/650 | 0.5638 | 0.5247 | 0.3274 |
| 6 | SNP 2 | p6600-rs12831105 | Intron 1 | T, 0.1188 | CC/CT/TT | 566/151/11 | 0.3570 | 0.2239 | | 571/152/10 | -0.5681 | 0.4412 | |
| 6 | SNP 3 | p7650-rs11615630 | Intron 1 | A, 0.0436 | GG/GA | 685/67 | 0.3382 | 0.4702 | | 695/68 | -2.4339 | 0.0359 | |
| 6 | SNP 4 | p10292-rs4765181 | Intron 1 | T, 0.2490 | TT/GT/GG | 44/285/417 | 0.1348 | 0.5405 | | 44/287/426 | -0.0286 | 0.9583 | |
| 7 | SNP 1 | p6600-rs12831105 | Intron 1 | T, 0.1188 | CC/CT/TT | 566/151/11 | 0.3570 | 0.2239 | 0.4139 | 571/152/10 | -0.5681 | 0.4412 | 0.1691 |
| 7 | SNP 2 | p7650-rs11615630 | Intron 1 | A, 0.0436 | GG/GA | 685/67 | 0.3382 | 0.4702 | | 695/68 | -2.4339 | 0.0359 | |
| 7 | SNP 3 | p10292-rs4765181 | Intron 1 | T, 0.2490 | TT/GT/GG | 44/285/417 | 0.1348 | 0.5405 | | 44/287/426 | -0.0286 | 0.9583 | |
| 7 | SNP 4 | p10991-rs10773112 | Intron 1 | A, 0.3534 | GG/GA/AA | 303/341/89 | 0.2146 | 0.2860 | | 309/339/90 | 0.4896 | 0.3230 | |
| 8 | SNP 1 | p7650-rs11615630 | Intron 1 | A, 0.0436 | GG/GA | 685/67 | 0.3382 | 0.4702 | 0.2071 | 695/68 | -2.4339 | 0.0359 | 0.1742 |
| 8 | SNP 2 | p10292-rs4765181 | Intron 1 | T, 0.2490 | TT/GT/GG | 44/285/417 | 0.1348 | 0.5405 | | 44/287/426 | -0.0286 | 0.9583 | |
| 8 | SNP 3 | p10991-rs10773112 | Intron 1 | A, 0.3534 | GG/GA/AA | 303/341/89 | 0.2146 | 0.2860 | | 309/339/90 | 0.4896 | 0.3230 | |
| 8 | SNP 4 | p13570-rs11057864 | Intron 1 | T, 0.1180 | TT/GT/GG | 14/147/581 | -0.0393 | 0.8917 | | 15/147/591 | 0.3006 | 0.6725 | |
| 9 | SNP 1 | p10292-rs4765181 | Intron 1 | T, 0.2490 | TT/GT/GG | 44/285/417 | 0.1348 | 0.5405 | 0.6128 | 44/287/426 | -0.0286 | 0.9583 | 0.5689 |
| 9 | SNP 2 | p10991-rs10773112 | Intron 1 | A, 0.3534 | GG/GA/AA | 303/341/89 | 0.2146 | 0.2860 | | 309/339/90 | 0.4896 | 0.3230 | |
| 9 | SNP 3 | p13570-rs11057864 | Intron 1 | T, 0.1180 | TT/GT/GG | 14/147/581 | -0.0393 | 0.8917 | | 15/147/591 | 0.3006 | 0.6725 | |
| 9 | SNP 4 | p16565-rs10773111 | Intron 1 | A, 0.1928 | GG/GA/AA | 481/239/24 | 0.1089 | 0.6550 | | 486/246/23 | 0.4484 | 0.4608 | |
| 10 | SNP 1 | p10991-rs10773112 | Intron 1 | A, 0.3534 | GG/GA/AA | 303/341/89 | 0.2146 | 0.2860 | 0.5086 | 309/339/90 | 0.4896 | 0.3230 | 0.8093 |
| 10 | SNP 2 | p13570-rs11057864 | Intron 1 | T, 0.1180 | TT/GT/GG | 14/147/581 | -0.0393 | 0.8917 | | 15/147/591 | 0.3006 | 0.6725 | |
| 10 | SNP 3 | p16565-rs10773111 | Intron 1 | A, 0.1928 | GG/GA/AA | 481/239/24 | 0.1089 | 0.6550 | | 486/246/23 | 0.4484 | 0.4608 | |
| 10 | SNP 4 | p20207-rs11057853 | Intron 1 | A, 0.4484 | AA/GA/GG | 150/360/228 | 0.4082 | 0.0343 | | 153/366/230 | 0.8422 | 0.0751 | |
| 11 | SNP 1 | p13570-rs11057864 | Intron 1 | T, 0.1180 | TT/GT/GG | 14/147/581 | -0.0393 | 0.8917 | 0.4147 | 15/147/591 | 0.3006 | 0.6725 | 0.4335 |
| 11 | SNP 2 | p16565-rs10773111 | Intron 1 | A, 0.1928 | GG/GA/AA | 481/239/24 | 0.1089 | 0.6550 | | 486/246/23 | 0.4484 | 0.4608 | |
| 11 | SNP 3 | p20207-rs11057853 | Intron 1 | A, 0.4484 | AA/GA/GG | 150/360/228 | 0.4082 | 0.0343 | | 153/366/230 | 0.8422 | 0.0751 | |
| 11 | SNP 4 | p20694-rs11057852 | Intron 1 | A, 0.1050 | GG/GA/AA | 581/130/11 | -0.0308 | 0.9198 | | 587/129/11 | 0.3519 | 0.6452 | |
| 12 | SNP 1 | p16565-rs10773111 | Intron 1 | A, 0.1928 | GG/GA/AA | 481/239/24 | 0.1089 | 0.6550 | 0.0764 | 486/246/23 | 0.4484 | 0.4608 | 0.2988 |
| 12 | SNP 2 | p20207-rs11057853 | Intron 1 | A, 0.4484 | AA/GA/GG | 150/360/228 | 0.4082 | 0.0343 | | 153/366/230 | 0.8422 | 0.0751 | |
| 12 | SNP 3 | p20694-rs11057852 | Intron 1 | A, 0.1050 | GG/GA/AA | 581/130/11 | -0.0308 | 0.9198 | | 587/129/11 | 0.3519 | 0.6452 | |
| 12 | SNP 4 | p20741-rs11057851 | Intron 1 | T, 0.3237 | TT/CT/CC | 72/334/328 | -0.5924 | 0.0043 | | 73/336/337 | -1.2331 | 0.0162 | |
| 13 | SNP 1 | p20207-rs11057853 | Intron 1 | A, 0.4484 | AA/GA/GG | 150/360/228 | 0.4082 | 0.0343 | 0.0762 | 153/366/230 | 0.8422 | 0.0751 | 0.0931 |
| 13 | SNP 2 | p20694-rs11057852 | Intron 1 | A, 0.1050 | GG/GA/AA | 581/130/11 | -0.0308 | 0.9198 | | 587/129/11 | 0.3519 | 0.6452 | |
| 13 | SNP 3 | p20741-rs11057851 | Intron 1 | T, 0.3237 | TT/CT/CC | 72/334/328 | -0.5924 | 0.0043 | | 73/336/337 | -1.2331 | 0.0162 | |
| 13 | SNP 4 | p21145-rs3924313 | Intron 1 | T, 0.1772 | CC/CT/TT | 503/219/22 | 0.1488 | 0.5502 | | 511/221/23 | 0.1215 | 0.8437 | |
| 14 | SNP 1 | p20694-rs11057852 | Intron 1 | A, 0.1050 | GG/GA/AA | 581/130/11 | -0.0308 | 0.9198 | 0.0886 | 587/129/11 | 0.3519 | 0.6452 | 0.1116 |
| 14 | SNP 2 | p20741-rs11057851 | Intron 1 | T, 0.3237 | TT/CT/CC | 72/334/328 | -0.5924 | 0.0043 | | 73/336/337 | -1.2331 | 0.0162 | |
| 14 | SNP 3 | p21145-rs3924313 | Intron 1 | T, 0.1772 | CC/CT/TT | 503/219/22 | 0.1488 | 0.5502 | | 511/221/23 | 0.1215 | 0.8437 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | SNP 4 | p22116-rs12370382 | Intron 1 | A, 0.0645 | GG/GA/AA | 643/86/3 | 0.1369 | 0.7300 | | 650/89/3 | 0.5264 | 0.5882 | |
| 15 | SNP 1 | p20741-rs11057851 | Intron 1 | T, 0.3237 | TT/CT/CC | 72/334/328 | -0.5924 | 0.0043 | 0.1004 | 73/336/337 | -1.2331 | 0.0162 | 0.1461 |
| 15 | SNP 2 | p21145-rs3924313 | Intron 1 | T, 0.1772 | CC/CT/TT | 503/219/22 | 0.1488 | 0.5502 | | 511/221/23 | 0.1215 | 0.8437 | |
| 15 | SNP 3 | p22116-rs12370382 | Intron 1 | A, 0.0645 | GG/GA/AA | 643/86/3 | 0.1369 | 0.7300 | | 650/89/3 | 0.5264 | 0.5882 | |
| 15 | SNP 4 | p22168-rs7137797 | Intron 1 | C, 0.3977 | CC/TC/TT | 113/355/259 | 0.2153 | 0.2763 | | 112/358/262 | 0.5066 | 0.3003 | |
| 16 | SNP 1 | p21145-rs3924313 | Intron 1 | T, 0.1772 | CC/CT/TT | 503/219/22 | 0.1488 | 0.5502 | 0.4920 | 511/221/23 | 0.1215 | 0.8437 | 0.7203 |
| 16 | SNP 2 | p22116-rs12370382 | Intron 1 | A, 0.0645 | GG/GA/AA | 643/86/3 | 0.1369 | 0.7300 | | 650/89/3 | 0.5264 | 0.5882 | |
| 16 | SNP 3 | p22168-rs7137797 | Intron 1 | C, 0.3977 | CC/TC/TT | 113/355/259 | 0.2153 | 0.2763 | | 112/358/262 | 0.5066 | 0.3003 | |
| 16 | SNP 4 | p22331-rs6488944 | Intron 1 | G, 0.1474 | TT/TG/GG | 522/188/12 | 0.3091 | 0.2661 | | 524/191/12 | 0.3684 | 0.5936 | |
| 17 | SNP 1 | p22116-rs12370382 | Intron 1 | A, 0.0645 | GG/GA/AA | 643/86/3 | 0.1369 | 0.7300 | 0.2834 | 650/89/3 | 0.5264 | 0.5882 | 0.6519 |
| 17 | SNP 2 | p22168-rs7137797 | Intron 1 | C, 0.3977 | CC/TC/TT | 113/355/259 | 0.2153 | 0.2763 | | 112/358/262 | 0.5066 | 0.3003 | |
| 17 | SNP 3 | p22331-rs6488944 | Intron 1 | G, 0.1474 | TT/TG/GG | 522/188/12 | 0.3091 | 0.2661 | | 524/191/12 | 0.3684 | 0.5936 | |
| 17 | SNP 4 | p22675-rs12425134 | Intron 1 | T, 0.0526 | TT/GT/GG | 2/75/662 | 0.7556 | 0.0738 | | 2/74/668 | 1.1151 | 0.2900 | |
| 18 | SNP 1 | p22168-rs7137797 | Intron 1 | C, 0.3977 | CC/TC/TT | 113/355/259 | 0.2153 | 0.2763 | 0.2862 | 112/358/262 | 0.5066 | 0.3003 | 0.7096 |
| 18 | SNP 2 | p22331-rs6488944 | Intron 1 | G, 0.1474 | TT/TG/GG | 522/188/12 | 0.3091 | 0.2661 | | 524/191/12 | 0.3684 | 0.5936 | |
| 18 | SNP 3 | p22675-rs12425134 | Intron 1 | T, 0.0526 | TT/GT/GG | 2/75/662 | 0.7556 | 0.0738 | | 2/74/668 | 1.1151 | 0.2900 | |
| 18 | SNP 4 | p28137-rs12229555 | Intron 1 | G, 0.3896 | GG/AG/AA | 99/373/257 | -0.3976 | 0.0500 | | 100/373/265 | -0.5752 | 0.2524 | |
| 19 | SNP 1 | p22331-rs6488944 | Intron 1 | G, 0.1474 | TT/TG/GG | 522/188/12 | 0.3091 | 0.2661 | 0.1757 | 524/191/12 | 0.3684 | 0.5936 | 0.5933 |
| 19 | SNP 2 | p22675-rs12425134 | Intron 1 | T, 0.0526 | TT/GT/GG | 2/75/662 | 0.7556 | 0.0738 | | 2/74/668 | 1.1151 | 0.2900 | |
| 19 | SNP 3 | p28137-rs12229555 | Intron 1 | G, 0.3896 | GG/AG/AA | 99/373/257 | -0.3976 | 0.0500 | | 100/373/265 | -0.5752 | 0.2524 | |
| 19 | SNP 4 | p28692-rs4765622 | Intron 1 | T, 0.2565 | TT/CT/CC | 43/289/403 | 0.1962 | 0.3817 | | 42/298/406 | 0.7871 | 0.1553 | |
| 20 | SNP 1 | p22675-rs12425134 | Intron 1 | T, 0.0526 | TT/GT/GG | 2/75/662 | 0.7556 | 0.0738 | 0.1080 | 2/74/668 | 1.1151 | 0.2900 | 0.2584 |
| 20 | SNP 2 | p28137-rs12229555 | Intron 1 | G, 0.3896 | GG/AG/AA | 99/373/257 | -0.3976 | 0.0500 | | 100/373/265 | -0.5752 | 0.2524 | |
| 20 | SNP 3 | p28692-rs4765622 | Intron 1 | T, 0.2565 | TT/CT/CC | 43/289/403 | 0.1962 | 0.3817 | | 42/298/406 | 0.7871 | 0.1553 | |
| 20 | SNP 4 | p28957-rs11057844 | Intron 1 | A, 0.2362 | GG/GA/AA | 428/263/40 | 0.3671 | 0.1075 | | 433/269/40 | 0.6107 | 0.2781 | |
| 21 | SNP 1 | p28137-rs12229555 | Intron 1 | G, 0.3896 | GG/AG/AA | 99/373/257 | -0.3976 | 0.0500 | 0.2221 | 100/373/265 | -0.5752 | 0.2524 | 0.2655 |
| 21 | SNP 2 | p28692-rs4765622 | Intron 1 | T, 0.2565 | TT/CT/CC | 43/289/403 | 0.1962 | 0.3817 | | 42/298/406 | 0.7871 | 0.1553 | |
| 21 | SNP 3 | p28957-rs11057844 | Intron 1 | A, 0.2362 | GG/GA/AA | 428/263/40 | 0.3671 | 0.1075 | | 433/269/40 | 0.6107 | 0.2781 | |
| 21 | SNP 4 | p29749-rs10846751 | Intron 1 | T, 0.4492 | TT/CT/CC | 142/365/210 | -0.3558 | 0.0701 | | 142/366/215 | -0.7763 | 0.1104 | |
| 22 | SNP 1 | p28692-rs4765622 | Intron 1 | T, 0.2565 | TT/CT/CC | 43/289/403 | 0.1962 | 0.3817 | 0.1369 | 42/298/406 | 0.7871 | 0.1553 | 0.6135 |
| 22 | SNP 2 | p28957-rs11057844 | Intron 1 | A, 0.2362 | GG/GA/AA | 428/263/40 | 0.3671 | 0.1075 | | 433/269/40 | 0.6107 | 0.2781 | |
| 22 | SNP 3 | p29749-rs10846751 | Intron 1 | T, 0.4492 | TT/CT/CC | 142/365/210 | -0.3558 | 0.0701 | | 142/366/215 | -0.7763 | 0.1104 | |
| 22 | SNP 4 | p31072-rs10846749 | Intron 1 | G, 0.4461 | CC/CG/GG | 222/366/144 | -0.3792 | 0.0510 | | 226/371/146 | -0.8005 | 0.0970 | |
| 23 | SNP 1 | p28957-rs11057844 | Intron 1 | A, 0.2362 | GG/GA/AA | 428/263/40 | 0.3671 | 0.1075 | 0.2085 | 433/269/40 | 0.6107 | 0.2781 | 0.6333 |
| 23 | SNP 2 | p29749-rs10846751 | Intron 1 | T, 0.4492 | TT/CT/CC | 142/365/210 | -0.3558 | 0.0701 | | 142/366/215 | -0.7763 | 0.1104 | |
| 23 | SNP 3 | p31072-rs10846749 | Intron 1 | G, 0.4461 | CC/CG/GG | 222/366/144 | -0.3792 | 0.0510 | | 226/371/146 | -0.8005 | 0.0970 | |
| 23 | SNP 4 | p31938-rs10744182 | Intron 1 | A, 0.1837 | AA/GA/GG | 25/221/487 | -0.2885 | 0.2438 | | 25/226/493 | -0.4389 | 0.4748 | |
| 24 | SNP 1 | p29749-rs10846751 | Intron 1 | T, 0.4492 | TT/CT/CC | 142/365/210 | -0.3558 | 0.0701 | 0.1804 | 142/366/215 | -0.7763 | 0.1104 | 0.5858 |
| 24 | SNP 2 | p31072-rs10846749 | Intron 1 | G, 0.4461 | CC/CG/GG | 222/366/144 | -0.3792 | 0.0510 | | 226/371/146 | -0.8005 | 0.0970 | |
| 24 | SNP 3 | p31938-rs10744182 | Intron 1 | A, 0.1837 | AA/GA/GG | 25/221/487 | -0.2885 | 0.2438 | | 25/226/493 | -0.4389 | 0.4748 | |
| 24 | SNP 4 | p32129-rs10773107 | Intron 1 | T, 0.1009 | TT/GT/GG | 9/132/593 | -0.3186 | 0.3107 | | 9/131/605 | 0.8714 | 0.2605 | |
| 25 | SNP 1 | p31072-rs10846749 | Intron 1 | G, 0.4461 | CC/CG/GG | 222/366/144 | -0.3792 | 0.0510 | 0.1155 | 226/371/146 | -0.8005 | 0.0970 | 0.3437 |
| 25 | SNP 2 | p31938-rs10744182 | Intron 1 | A, 0.1837 | AA/GA/GG | 25/221/487 | -0.2885 | 0.2438 | | 25/226/493 | -0.4389 | 0.4748 | |
| 25 | SNP 3 | p32129-rs10773107 | Intron 1 | T, 0.1009 | TT/GT/GG | 9/132/593 | -0.3186 | 0.3107 | | 9/131/605 | 0.8714 | 0.2605 | |
| 25 | SNP 4 | p32273-rs12580803 | Intron 1 | C, 0.1006 | TT/TC/CC | 619/114/18 | 0.2010 | 0.4984 | | 627/117/18 | 1.2550 | 0.0842 | |
| 26 | SNP 1 | p31938-rs10744182 | Intron 1 | A, 0.1837 | AA/GA/GG | 25/221/487 | -0.2885 | 0.2438 | 0.9308 | 25/226/493 | -0.4389 | 0.4748 | 0.1939 |
| 26 | SNP 2 | p32129-rs10773107 | Intron 1 | T, 0.1009 | TT/GT/GG | 9/132/593 | -0.3186 | 0.3107 | | 9/131/605 | 0.8714 | 0.2605 | |
| 26 | SNP 3 | p32273-rs12580803 | Intron 1 | C, 0.1006 | TT/TC/CC | 619/114/18 | 0.2010 | 0.4984 | | 627/117/18 | 1.2550 | 0.0842 | |
| 26 | SNP 4 | p32290-rs10744181 | Intron 1 | C, 0.1238 | TT/TC/CC | 547/141/18 | -0.1852 | 0.5162 | | 551/143/18 | 0.1584 | 0.8211 | |
| 27 | SNP 1 | p32129-rs10773107 | Intron 1 | T, 0.1009 | TT/GT/GG | 9/132/593 | -0.3186 | 0.3107 | 0.7762 | 9/131/605 | 0.8714 | 0.2605 | 0.5428 |
| 27 | SNP 2 | p32273-rs12580803 | Intron 1 | C, 0.1006 | TT/TC/CC | 619/114/18 | 0.2010 | 0.4984 | | 627/117/18 | 1.2550 | 0.0842 | |
| 27 | SNP 3 | p32290-rs10744181 | Intron 1 | C, 0.1238 | TT/TC/CC | 547/141/18 | -0.1852 | 0.5162 | | 551/143/18 | 0.1584 | 0.8211 | |
| 27 | SNP 4 | p32395-rs12581963 | Intron 1 | T, 0.1314 | TT/CT/CC | 11/179/557 | -0.3173 | 0.2589 | | 11/177/570 | -0.6483 | 0.3556 | |
| 28 | SNP 1 | p32273-rs12580803 | Intron 1 | C, 0.1006 | TT/TC/CC | 619/114/18 | 0.2010 | 0.4984 | 0.6853 | 627/117/18 | 1.2550 | 0.0842 | 0.8033 |
| 28 | SNP 2 | p32290-rs10744181 | Intron 1 | C, 0.1238 | TT/TC/CC | 547/141/18 | -0.1852 | 0.5162 | | 551/143/18 | 0.1584 | 0.8211 | |
| 28 | SNP 3 | p32395-rs12581963 | Intron 1 | T, 0.1314 | TT/CT/CC | 11/179/557 | -0.3173 | 0.2589 | | 11/177/570 | -0.6483 | 0.3556 | |
| 28 | SNP 4 | p32750-rs7967521 | Intron 1 | G, 0.3425 | GG/AG/AA | 89/306/310 | -0.2137 | 0.2936 | | 91/304/315 | 0.0564 | 0.9104 | |
| 29 | SNP 1 | p32290-rs10744181 | Intron 1 | C, 0.1238 | TT/TC/CC | 547/141/18 | -0.1852 | 0.5162 | 0.3363 | 551/143/18 | 0.1584 | 0.8211 | 0.8929 |
| 29 | SNP 2 | p32395-rs12581963 | Intron 1 | T, 0.1314 | TT/CT/CC | 11/179/557 | -0.3173 | 0.2589 | | 11/177/570 | -0.6483 | 0.3556 | |
| 29 | SNP 3 | p32750-rs7967521 | Intron 1 | G, 0.3425 | GG/AG/AA | 89/306/310 | -0.2137 | 0.2936 | | 91/304/315 | 0.0564 | 0.9104 | |
| 29 | SNP 4 | p32777-rs11057841 | Intron 1 | A, 0.2805 | GG/GA/AA | 367/307/50 | -0.1217 | 0.5781 | | 374/303/52 | 0.0373 | 0.9450 | |
| 30 | SNP 1 | p32395-rs12581963 | Intron 1 | T, 0.1314 | TT/CT/CC | 11/179/557 | -0.3173 | 0.2589 | 0.3979 | 11/177/570 | -0.6483 | 0.3556 | 0.9607 |
| 30 | SNP 2 | p32750-rs7967521 | Intron 1 | G, 0.3425 | GG/AG/AA | 89/306/310 | -0.2137 | 0.2936 | | 91/304/315 | 0.0564 | 0.9104 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 30 | SNP 3 | p32777-rs11057841 | Intron 1 | A, 0.2805 | GG/GA/AA | 367/307/50 | -0.1217 | 0.5781 | | 374/303/52 | 0.0373 | 0.9450 | |
| 30 | SNP 4 | p32860-rs7967406 | Intron 1 | C, 0.0991 | CC/AC/AA | 9/131/608 | 0.2647 | 0.3996 | | 9/135/616 | 0.4768 | 0.5369 | |
| 31 | SNP 1 | p32750-rs7967521 | Intron 1 | G, 0.3425 | GG/AG/AA | 89/306/310 | -0.2137 | 0.2936 | 0.3062 | 91/304/315 | 0.0564 | 0.9104 | 0.9880 |
| 31 | SNP 2 | p32777-rs11057841 | Intron 1 | A, 0.2805 | GG/GA/AA | 367/307/50 | -0.1217 | 0.5781 | | 374/303/52 | 0.0373 | 0.9450 | |
| 31 | SNP 3 | p32860-rs7967406 | Intron 1 | C, 0.0991 | CC/AC/AA | 9/131/608 | 0.2647 | 0.3996 | | 9/135/616 | 0.4768 | 0.5369 | |
| 31 | SNP 4 | p33531-rs11057838 | Intron 1 | A, 0.2278 | CC/CA/AA | 439/254/41 | -0.2064 | 0.3609 | | 440/259/40 | 0.1993 | 0.7218 | |
| 32 | SNP 1 | p32777-rs11057841 | Intron 1 | A, 0.2805 | GG/GA/AA | 367/307/50 | -0.1217 | 0.5781 | 0.3039 | 374/303/52 | 0.0373 | 0.9450 | 0.9957 |
| 32 | SNP 2 | p32860-rs7967406 | Intron 1 | C, 0.0991 | CC/AC/AA | 9/131/608 | 0.2647 | 0.3996 | | 9/135/616 | 0.4768 | 0.5369 | |
| 32 | SNP 3 | p33531-rs11057838 | Intron 1 | A, 0.2278 | CC/CA/AA | 439/254/41 | -0.2064 | 0.3609 | | 440/259/40 | 0.1993 | 0.7218 | |
| 32 | SNP 4 | p36094-rs11608336 | Intron 1 | A, 0.1543 | GG/GA/AA | 521/195/15 | 0.3354 | 0.2108 | | 526/195/16 | -0.0956 | 0.8848 | |
| 33 | SNP 1 | p32860-rs7967406 | Intron 1 | C, 0.0991 | CC/AC/AA | 9/131/608 | 0.2647 | 0.3996 | 0.1952 | 9/135/616 | 0.4768 | 0.5369 | 0.5389 |
| 33 | SNP 2 | p33531-rs11057838 | Intron 1 | A, 0.2278 | CC/CA/AA | 439/254/41 | -0.2064 | 0.3609 | | 440/259/40 | 0.1993 | 0.7218 | |
| 33 | SNP 3 | p36094-rs11608336 | Intron 1 | A, 0.1543 | GG/GA/AA | 521/195/15 | 0.3354 | 0.2108 | | 526/195/16 | -0.0956 | 0.8848 | |
| 33 | SNP 4 | p36361-rs4765178 | Intron 1 | T, 0.1671 | TT/CT/CC | 19/201/496 | -0.1369 | 0.6024 | | 18/205/497 | 0.5087 | 0.4368 | |
| 34 | SNP 1 | p33531-rs11057838 | Intron 1 | A, 0.2278 | CC/CA/AA | 439/254/41 | -0.2064 | 0.3609 | 0.7481 | 440/259/40 | 0.1993 | 0.7218 | 0.3649 |
| 34 | SNP 2 | p36094-rs11608336 | Intron 1 | A, 0.1543 | GG/GA/AA | 521/195/15 | 0.3354 | 0.2108 | | 526/195/16 | -0.0956 | 0.8848 | |
| 34 | SNP 3 | p36361-rs4765178 | Intron 1 | T, 0.1671 | TT/CT/CC | 19/201/496 | -0.1369 | 0.6024 | | 18/205/497 | 0.5087 | 0.4368 | |
| 34 | SNP 4 | p36908-rs10846745 | Intron 1 | G, 0.3257 | GG/CG/CC | 76/334/329 | -0.0384 | 0.8511 | | 78/335/337 | 0.0089 | 0.9859 | |
| 35 | SNP 1 | p36094-rs11608336 | Intron 1 | A, 0.1543 | GG/GA/AA | 521/195/15 | 0.3354 | 0.2108 | 0.9219 | 526/195/16 | -0.0956 | 0.8848 | 0.8447 |
| 35 | SNP 2 | p36361-rs4765178 | Intron 1 | T, 0.1671 | TT/CT/CC | 19/201/496 | -0.1369 | 0.6024 | | 18/205/497 | 0.5087 | 0.4368 | |
| 35 | SNP 3 | p36908-rs10846745 | Intron 1 | G, 0.3257 | GG/CG/CC | 76/334/329 | -0.0384 | 0.8511 | | 78/335/337 | 0.0089 | 0.9859 | |
| 35 | SNP 4 | p37095-rs10846744 | Intron 1 | G, 0.3056 | CC/CG/GG | 348/340/57 | 0.1168 | 0.5829 | | 350/350/56 | 0.1538 | 0.7732 | |
| 36 | SNP 1 | p36361-rs4765178 | Intron 1 | T, 0.1671 | TT/CT/CC | 19/201/496 | -0.1369 | 0.6024 | 0.4949 | 18/205/497 | 0.5087 | 0.4368 | 0.9787 |
| 36 | SNP 2 | p36908-rs10846745 | Intron 1 | G, 0.3257 | GG/CG/CC | 76/334/329 | -0.0384 | 0.8511 | | 78/335/337 | 0.0089 | 0.9859 | |
| 36 | SNP 3 | p37095-rs10846744 | Intron 1 | G, 0.3056 | CC/CG/GG | 348/340/57 | 0.1168 | 0.5829 | | 350/350/56 | 0.1538 | 0.7732 | |
| 36 | SNP 4 | p41632-rs6488943 | Intron 1 | C, 0.2954 | CC/AC/AA | 50/309/337 | -0.2195 | 0.3244 | | 52/312/340 | 0.3025 | 0.5783 | |
| 37 | SNP 1 | p36908-rs10846745 | Intron 1 | G, 0.3257 | GG/CG/CC | 76/334/329 | -0.0384 | 0.8511 | 0.8624 | 78/335/337 | 0.0089 | 0.9859 | 0.7511 |
| 37 | SNP 2 | p37095-rs10846744 | Intron 1 | G, 0.3056 | CC/CG/GG | 348/340/57 | 0.1168 | 0.5829 | | 350/350/56 | 0.1538 | 0.7732 | |
| 37 | SNP 3 | p41632-rs6488943 | Intron 1 | C, 0.2954 | CC/AC/AA | 50/309/337 | -0.2195 | 0.3244 | | 52/312/340 | 0.3025 | 0.5783 | |
| 37 | SNP 4 | p42467-rs11057830 | Intron 1 | T, 0.1523 | TT/CT/CC | 13/201/523 | -0.2810 | 0.3015 | | 14/200/533 | -0.9351 | 0.1612 | |
| 38 | SNP 1 | p37095-rs10846744 | Intron 1 | G, 0.3056 | CC/CG/GG | 348/340/57 | 0.1168 | 0.5829 | 0.4622 | 350/350/56 | 0.1538 | 0.7732 | 0.8715 |
| 38 | SNP 2 | p41632-rs6488943 | Intron 1 | C, 0.2954 | CC/AC/AA | 50/309/337 | -0.2195 | 0.3244 | | 52/312/340 | 0.3025 | 0.5783 | |
| 38 | SNP 3 | p42467-rs11057830 | Intron 1 | T, 0.1523 | TT/CT/CC | 13/201/523 | -0.2810 | 0.3015 | | 14/200/533 | -0.9351 | 0.1612 | |
| 38 | SNP 4 | p45516-rs1902569 | Intron 1 | A, 0.1544 | AA/GA/GG | 18/190/519 | 0.5447 | 0.0386 | | 18/191/523 | 0.5127 | 0.4331 | |
| 39 | SNP 1 | p41632-rs6488943 | Intron 1 | C, 0.2954 | CC/AC/AA | 50/309/337 | -0.2195 | 0.3244 | **0.0207** | 52/312/340 | 0.3025 | 0.5783 | 0.4914 |
| 39 | SNP 2 | p42467-rs11057830 | Intron 1 | T, 0.1523 | TT/CT/CC | 13/201/523 | -0.2810 | 0.3015 | | 14/200/533 | -0.9351 | 0.1612 | |
| 39 | SNP 3 | p45516-rs1902569 | Intron 1 | A, 0.1544 | AA/GA/GG | 18/190/519 | 0.5447 | 0.0386 | | 18/191/523 | 0.5127 | 0.4331 | |
| 39 | SNP 4 | p45627-rs12297372 | Intron 1 | G, 0.0487 | GG/AG/AA | 1/68/659 | -0.0483 | 0.9156 | | 1/71/661 | 0.9720 | 0.3816 | |
| 40 | SNP 1 | p42467-rs11057830 | Intron 1 | T, 0.1523 | TT/CT/CC | 13/201/523 | -0.2810 | 0.3015 | 0.3346 | 14/200/533 | -0.9351 | 0.1612 | 0.5468 |
| 40 | SNP 2 | p45516-rs1902569 | Intron 1 | A, 0.1544 | AA/GA/GG | 18/190/519 | 0.5447 | 0.0386 | | 18/191/523 | 0.5127 | 0.4331 | |
| 40 | SNP 3 | p45627-rs12297372 | Intron 1 | G, 0.0487 | GG/AG/AA | 1/68/659 | -0.0483 | 0.9156 | | 1/71/661 | 0.9720 | 0.3816 | |
| 40 | SNP 4 | p46964-rs114061302 | Intron 1 | A, 0.0388 | AA/GA/GG | 1/55/688 | 0.0526 | 0.9158 | | 1/58/696 | 0.4839 | 0.6895 | |
| 41 | SNP 1 | p45516-rs1902569 | Intron 1 | A, 0.1544 | AA/GA/GG | 18/190/519 | 0.5447 | 0.0386 | 0.2665 | 18/191/523 | 0.5127 | 0.4331 | 0.8055 |
| 41 | SNP 2 | p45627-rs12297372 | Intron 1 | G, 0.0487 | GG/AG/AA | 1/68/659 | -0.0483 | 0.9156 | | 1/71/661 | 0.9720 | 0.3816 | |
| 41 | SNP 3 | p46964-rs114061302 | Intron 1 | A, 0.0388 | AA/GA/GG | 1/55/688 | 0.0526 | 0.9158 | | 1/58/696 | 0.4839 | 0.6895 | |
| 41 | SNP 4 | p48969-rs2343394 | Intron 2 | T, 0.1898 | TT/CT/CC | 32/225/491 | 0.3165 | 0.1788 | | 32/226/501 | 0.6411 | 0.2714 | |
| 42 | SNP 1 | p45627-rs12297372 | Intron 1 | G, 0.0487 | GG/AG/AA | 1/68/659 | -0.0483 | 0.9156 | 0.6060 | 1/71/661 | 0.9720 | 0.3816 | 0.8644 |
| 42 | SNP 2 | p46964-rs114061302 | Intron 1 | A, 0.0388 | AA/GA/GG | 1/55/688 | 0.0526 | 0.9158 | | 1/58/696 | 0.4839 | 0.6895 | |
| 42 | SNP 3 | p48969-rs2343394 | Intron 2 | T, 0.1898 | TT/CT/CC | 32/225/491 | 0.3165 | 0.1788 | | 32/226/501 | 0.6411 | 0.2714 | |
| 42 | SNP 4 | p49537-rs7305310 | Intron 2 | T, 0.1007 | CC/CT/TT | 595/117/16 | -0.3396 | 0.2566 | | 601/118/16 | -0.2265 | 0.7599 | |
| 43 | SNP 1 | p46964-rs114061302 | Intron 1 | A, 0.0388 | AA/GA/GG | 1/55/688 | 0.0526 | 0.9158 | 0.2986 | 1/58/696 | 0.4839 | 0.6895 | 0.6379 |
| 43 | SNP 2 | p48969-rs2343394 | Intron 2 | T, 0.1898 | TT/CT/CC | 32/225/491 | 0.3165 | 0.1788 | | 32/226/501 | 0.6411 | 0.2714 | |
| 43 | SNP 3 | p49537-rs7305310 | Intron 2 | T, 0.1007 | CC/CT/TT | 595/117/16 | -0.3396 | 0.2566 | | 601/118/16 | -0.2265 | 0.7599 | |
| 43 | SNP 4 | p49570delC-rs145376237 | Intron 2 | delC, 0.2276 | DD/WD/WW | 36/260/432 | 0.3121 | 0.1773 | | 37/260/437 | 0.4929 | 0.3880 | |
| 44 | SNP 1 | p48969-rs2343394 | Intron 2 | T, 0.1898 | TT/CT/CC | 32/225/491 | 0.3165 | 0.1788 | **0.0271** | 32/226/501 | 0.6411 | 0.2714 | 0.3331 |
| 44 | SNP 2 | p49537-rs7305310 | Intron 2 | T, 0.1007 | CC/CT/TT | 595/117/16 | -0.3396 | 0.2566 | | 601/118/16 | -0.2265 | 0.7599 | |
| 44 | SNP 3 | p49570delC-rs145376237 | Intron 2 | delC, 0.2276 | DD/WD/WW | 36/260/432 | 0.3121 | 0.1773 | | 37/260/437 | 0.4929 | 0.3880 | |
| 44 | SNP 4 | p49690-rs4765615 | Intron 2 | A, 0.4426 | AA/GA/GG | 156/318/244 | -0.4646 | 0.0130 | | 157/323/244 | -0.9139 | 0.0480 | |
| 45 | SNP 1 | p49537-rs7305310 | Intron 2 | T, 0.1007 | CC/CT/TT | 595/117/16 | -0.3396 | 0.2566 | **0.0155** | 601/118/16 | -0.2265 | 0.7599 | 0.2586 |
| 45 | SNP 2 | p49570delC-rs145376237 | Intron 2 | delC, 0.2276 | DD/WD/WW | 36/260/432 | 0.3121 | 0.1773 | | 37/260/437 | 0.4929 | 0.3880 | |
| 45 | SNP 3 | p49690-rs4765615 | Intron 2 | A, 0.4426 | AA/GA/GG | 156/318/244 | -0.4646 | 0.0130 | | 157/323/244 | -0.9139 | 0.0480 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 45 | SNP 4 | p49759-rs146272788 | Intron 2 | T, 0.0020 | CC/CT | 725/3 | 2.5988 | 0.2219 | | 731/3 | 1.5883 | 0.7630 | |
| 46 | SNP 1 | p49570delC-rs145376237 | Intron 2 | delC, 0.2276 | DD/WD/WW | 36/260/432 | 0.3121 | 0.1773 | **0.0278** | 37/260/437 | 0.4929 | 0.3880 | 0.0675 |
| 46 | SNP 2 | p49690-rs4765615 | Intron 2 | A, 0.4426 | AA/GA/GG | 156/318/244 | -0.4646 | 0.0130 | | 157/323/244 | -0.9139 | 0.0480 | |
| 46 | SNP 3 | p49759-rs146272788 | Intron 2 | T, 0.0020 | CC/CT | 725/3 | 2.5988 | 0.2219 | | 731/3 | 1.5883 | 0.7630 | |
| 46 | SNP 4 | p49978-rs5891 | Exon 3 | A, 0.0058 | GA/GG | 9/743 | 1.3374 | 0.2791 | | 9/754 | 5.6762 | 0.0628 | |
| 47 | SNP 1 | p49690-rs4765615 | Intron 2 | A, 0.4426 | AA/GA/GG | 156/318/244 | -0.4646 | 0.0130 | **0.0079** | 157/323/244 | -0.9139 | 0.0480 | **0.0343** |
| 47 | SNP 2 | p49759-rs146272788 | Intron 2 | T, 0.0020 | CC/CT | 725/3 | 2.5988 | 0.2219 | | 731/3 | 1.5883 | 0.7630 | |
| 47 | SNP 3 | p49978-rs5891 | Exon 3 | A, 0.0058 | GA/GG | 9/743 | 1.3374 | 0.2791 | | 9/754 | 5.6762 | 0.0628 | |
| 47 | SNP 4 | p50024-rs368880622 | Intron 3 | T, 0.0026 | GG/GT | 737/3 | 1.6506 | 0.4362 | | 742/4 | 1.6012 | 0.7255 | |
| 48 | SNP 1 | p49759-rs146272788 | Intron 2 | T, 0.0020 | CC/CT | 725/3 | 2.5988 | 0.2219 | 0.1075 | 731/3 | 1.5883 | 0.7630 | **0.0293** |
| 48 | SNP 2 | p49978-rs5891 | Exon 3 | A, 0.0058 | GA/GG | 9/743 | 1.3374 | 0.2791 | | 9/754 | 5.6762 | 0.0628 | |
| 48 | SNP 3 | p50024-rs368880622 | Intron 3 | T, 0.0026 | GG/GT | 737/3 | 1.6506 | 0.4362 | | 742/4 | 1.6012 | 0.7255 | |
| 48 | SNP 4 | p50118-rs58710319 | Intron 3 | T, 0.0208 | CT/CC | 31/711 | 0.7885 | 0.2399 | | 32/719 | 3.1376 | 0.0571 | |
| 49 | SNP 1 | p49978-rs5891 | Exon 3 | A, 0.0058 | GA/GG | 9/743 | 1.3374 | 0.2791 | 0.0672 | 9/754 | 5.6762 | 0.0628 | **0.0289** |
| 49 | SNP 2 | p50024-rs368880622 | Intron 3 | T, 0.0026 | GG/GT | 737/3 | 1.6506 | 0.4362 | | 742/4 | 1.6012 | 0.7255 | |
| 49 | SNP 3 | p50118-rs58710319 | Intron 3 | T, 0.0208 | CT/CC | 31/711 | 0.7885 | 0.2399 | | 32/719 | 3.1376 | 0.0571 | |
| 49 | SNP 4 | p50151-rs2278986 | Intron 3 | C, 0.1933 | CC/TC/TT | 33/225/484 | 0.4333 | 0.0656 | | 33/226/494 | 0.8568 | 0.1419 | |
| 50 | SNP 1 | p50024-rs368880622 | Intron 3 | T, 0.0026 | GG/GT | 737/3 | 1.6506 | 0.4362 | 0.1980 | 742/4 | 1.6012 | 0.7255 | 0.1897 |
| 50 | SNP 2 | p50118-rs58710319 | Intron 3 | T, 0.0208 | CT/CC | 31/711 | 0.7885 | 0.2399 | | 32/719 | 3.1376 | 0.0571 | |
| 50 | SNP 3 | p50151-rs2278986 | Intron 3 | C, 0.1933 | CC/TC/TT | 33/225/484 | 0.4333 | 0.0656 | | 33/226/494 | 0.8568 | 0.1419 | |
| 50 | SNP 4 | p50380-rs141748317 | Intron 3 | G, 0.0112 | AA/AG | 723/15 | -0.0273 | 0.9772 | | 727/17 | 0.3920 | 0.8609 | |
| 51 | SNP 1 | p50118-rs58710319 | Intron 3 | T, 0.0208 | CT/CC | 31/711 | 0.7885 | 0.2399 | 0.1254 | 32/719 | 3.1376 | 0.0571 | 0.0779 |
| 51 | SNP 2 | p50151-rs2278986 | Intron 3 | C, 0.1933 | CC/TC/TT | 33/225/484 | 0.4333 | 0.0656 | | 33/226/494 | 0.8568 | 0.1419 | |
| 51 | SNP 3 | p50380-rs141748317 | Intron 3 | G, 0.0112 | AA/AG | 723/15 | -0.0273 | 0.9772 | | 727/17 | 0.3920 | 0.8609 | |
| 51 | SNP 4 | p50489-rs61320152 | Intron 3 | T, 0.0257 | GG/GT | 699/38 | 0.2390 | 0.6958 | | 704/39 | 2.1813 | 0.1439 | |
| 52 | SNP 1 | p50151-rs2278986 | Intron 3 | C, 0.1933 | CC/TC/TT | 33/225/484 | 0.4333 | 0.0656 | 0.2903 | 33/226/494 | 0.8568 | 0.1419 | 0.1905 |
| 52 | SNP 2 | p50380-rs141748317 | Intron 3 | G, 0.0112 | AA/AG | 723/15 | -0.0273 | 0.9772 | | 727/17 | 0.3920 | 0.8609 | |
| 52 | SNP 3 | p50489-rs61320152 | Intron 3 | T, 0.0257 | GG/GT | 699/38 | 0.2390 | 0.6958 | | 704/39 | 2.1813 | 0.1439 | |
| 52 | SNP 4 | p50954-chr12_125298566 | Intron 4 | C, 0.0007 | TC/TT | 1/735 | 4.5639 | 0.2131 | | 1/742 | -8.7626 | 0.3375 | |
| 53 | SNP 1 | p50380-rs141748317 | Intron 3 | G, 0.0112 | AA/AG | 723/15 | -0.0273 | 0.9772 | 0.9605 | 727/17 | 0.3920 | 0.8609 | 0.5439 |
| 53 | SNP 2 | p50489-rs61320152 | Intron 3 | T, 0.0257 | GG/GT | 699/38 | 0.2390 | 0.6958 | | 704/39 | 2.1813 | 0.1439 | |
| 53 | SNP 3 | p50954-chr12_125298566 | Intron 4 | C, 0.0007 | TC/TT | 1/735 | 4.5639 | 0.2131 | | 1/742 | -8.7626 | 0.3375 | |
| 53 | SNP 4 | p51888-rs7138304 | Intron 4 | T, 0.1079 | TT/CT/CC | 16/129/589 | -0.0610 | 0.8334 | | 16/130/593 | -0.0031 | 0.9965 | |
| 54 | SNP 1 | p50489-rs61320152 | Intron 3 | T, 0.0257 | GG/GT | 699/38 | 0.2390 | 0.6958 | 0.8543 | 704/39 | 2.1813 | 0.1439 | 0.6688 |
| 54 | SNP 2 | p50954-chr12_125298566 | Intron 4 | C, 0.0007 | TC/TT | 1/735 | 4.5639 | 0.2131 | | 1/742 | -8.7626 | 0.3375 | |
| 54 | SNP 3 | p51888-rs7138304 | Intron 4 | T, 0.1079 | TT/CT/CC | 16/129/589 | -0.0610 | 0.8334 | | 16/130/593 | -0.0031 | 0.9965 | |
| 54 | SNP 4 | p52096-rs10846739 | Intron 4 | G, 0.4693 | GG/AG/AA | 169/343/217 | 0.0195 | 0.9168 | | 172/342/221 | -0.1537 | 0.7377 | |
| 55 | SNP 1 | p50954-chr12_125298566 | Intron 4 | C, 0.0007 | TC/TT | 1/735 | 4.5639 | 0.2131 | 0.6351 | 1/742 | -8.7626 | 0.3375 | 0.5383 |
| 55 | SNP 2 | p51888-rs7138304 | Intron 4 | T, 0.1079 | TT/CT/CC | 16/129/589 | -0.0610 | 0.8334 | | 16/130/593 | -0.0031 | 0.9965 | |
| 55 | SNP 3 | p52096-rs10846739 | Intron 4 | G, 0.4693 | GG/AG/AA | 169/343/217 | 0.0195 | 0.9168 | | 172/342/221 | -0.1537 | 0.7377 | |
| 55 | SNP 4 | p52556-rs11057820 | Intron 4 | A, 0.1000 | AA/GA/GG | 12/124/610 | -0.1513 | 0.6235 | | 12/126/619 | -1.1588 | 0.1282 | |
| 56 | SNP 1 | p51888-rs7138304 | Intron 4 | T, 0.1079 | TT/CT/CC | 16/129/589 | -0.0610 | 0.8334 | 0.9587 | 16/130/593 | -0.0031 | 0.9965 | 0.3509 |
| 56 | SNP 2 | p52096-rs10846739 | Intron 4 | G, 0.4693 | GG/AG/AA | 169/343/217 | 0.0195 | 0.9168 | | 172/342/221 | -0.1537 | 0.7377 | |
| 56 | SNP 3 | p52556-rs11057820 | Intron 4 | A, 0.1000 | AA/GA/GG | 12/124/610 | -0.1513 | 0.6235 | | 12/126/619 | -1.1588 | 0.1282 | |
| 56 | SNP 4 | p52610-rs10846738 | Intron 4 | T, 0.1349 | TT/CT/CC | 12/172/545 | -0.0102 | 0.9710 | | 13/172/553 | -0.6490 | 0.3462 | |
| 57 | SNP 1 | p52096-rs10846739 | Intron 4 | G, 0.4693 | GG/AG/AA | 169/343/217 | 0.0195 | 0.9168 | 0.5802 | 172/342/221 | -0.1537 | 0.7377 | 0.3474 |
| 57 | SNP 2 | p52556-rs11057820 | Intron 4 | A, 0.1000 | AA/GA/GG | 12/124/610 | -0.1513 | 0.6235 | | 12/126/619 | -1.1588 | 0.1282 | |
| 57 | SNP 3 | p52610-rs10846738 | Intron 4 | T, 0.1349 | TT/CT/CC | 12/172/545 | -0.0102 | 0.9710 | | 13/172/553 | -0.6490 | 0.3462 | |
| 57 | SNP 4 | p52919-chr12_125296601 | Intron 4 | T, 0.0013 | GG/GT | 734/2 | -7.4063 | 0.0043 | | 741/2 | -13.4137 | 0.0385 | |
| 58 | SNP 1 | p52556-rs11057820 | Intron 4 | A, 0.1000 | AA/GA/GG | 12/124/610 | -0.1513 | 0.6235 | 0.2618 | 12/126/619 | -1.1588 | 0.1282 | 0.1625 |
| 58 | SNP 2 | p52610-rs10846738 | Intron 4 | T, 0.1349 | TT/CT/CC | 12/172/545 | -0.0102 | 0.9710 | | 13/172/553 | -0.6490 | 0.3462 | |
| 58 | SNP 3 | p52919-chr12_125296601 | Intron 4 | T, 0.0013 | GG/GT | 734/2 | -7.4063 | 0.0043 | | 741/2 | -13.4137 | 0.0385 | |
| 58 | SNP 4 | p52956-rs77740046 | Intron 4 | T, 0.0546 | CC/CT/TT | 651/75/3 | 0.3300 | 0.4288 | | 657/75/3 | -0.4428 | 0.6689 | |
| 59 | SNP 1 | p52610-rs10846738 | Intron 4 | T, 0.1349 | TT/CT/CC | 12/172/545 | -0.0102 | 0.9710 | 0.1054 | 13/172/553 | -0.6490 | 0.3462 | 0.4723 |
| 59 | SNP 2 | p52919-chr12_125296601 | Intron 4 | T, 0.0013 | GG/GT | 734/2 | -7.4063 | 0.0043 | | 741/2 | -13.4137 | 0.0385 | |
| 59 | SNP 3 | p52956-rs77740046 | Intron 4 | T, 0.0546 | CC/CT/TT | 651/75/3 | 0.3300 | 0.4288 | | 657/75/3 | -0.4428 | 0.6689 | |
| 59 | SNP 4 | p52995-rs113910315 | Intron 4-splice site | G, 0.0020 | TG/TT | 3/740 | -1.4175 | 0.5038 | | 3/746 | 0.6563 | 0.9010 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 60 | SNP 1 | p52919-chr12_125296601 | Intron 4 | T, 0.0013 | GG/GT | 734/2 | -7.4063 | 0.0043 | 0.1743 | 741/2 | -13.4137 | 0.0385 | 0.5102 |
| 60 | SNP 2 | p52956-rs77740046 | Intron 4 | T, 0.0546 | CC/CT/TT | 651/75/3 | 0.3300 | 0.4288 | | 657/75/3 | -0.4428 | 0.6689 | |
| 60 | SNP 3 | p52995-rs113910315 | Intron 4-splice site | G, 0.0020 | TG/TT | 3/740 | -1.4175 | 0.5038 | | 3/746 | 0.6563 | 0.9010 | |
| 60 | SNP 4 | p53359-rs112371713 | Intron 5 | A, 0.1243 | AA/GA/GG | 9/160/549 | 0.4193 | 0.1651 | | 10/161/554 | 0.8266 | 0.2605 | |
| 61 | SNP 1 | p52956-rs77740046 | Intron 4 | T, 0.0546 | CC/CT/TT | 651/75/3 | 0.3300 | 0.4288 | 0.1406 | 657/75/3 | -0.4428 | 0.6689 | 0.2847 |
| 61 | SNP 2 | p52995-rs113910315 | Intron 4-splice site | G, 0.0020 | TG/TT | 3/740 | -1.4175 | 0.5038 | | 3/746 | 0.6563 | 0.9010 | |
| 61 | SNP 3 | p53359-rs112371713 | Intron 5 | A, 0.1243 | AA/GA/GG | 9/160/549 | 0.4193 | 0.1651 | | 10/161/554 | 0.8266 | 0.2605 | |
| 61 | SNP 4 | p53372-rs115604379 | Intron 5 | T, 0.0066 | CC/CT | 729/10 | 3.0372 | 0.0093 | | 735/10 | 4.3130 | 0.1386 | |
| 62 | SNP 1 | p52995-rs113910315 | Intron 4-splice site | G, 0.0020 | TG/TT | 3/740 | -1.4175 | 0.5038 | 0.0712 | 3/746 | 0.6563 | 0.9010 | 0.3287 |
| 62 | SNP 2 | p53359-rs112371713 | Intron 5 | A, 0.1243 | AA/GA/GG | 9/160/549 | 0.4193 | 0.1651 | | 10/161/554 | 0.8266 | 0.2605 | |
| 62 | SNP 3 | p53372-rs115604379 | Intron 5 | T, 0.0066 | CC/CT | 729/10 | 3.0372 | 0.0093 | | 735/10 | 4.3130 | 0.1386 | |
| 62 | SNP 4 | p53790-rs4765614 | Intron 5 | A, 0.2653 | GG/GA/AA | 399/276/58 | -0.3281 | 0.1218 | | 402/277/59 | -0.4621 | 0.3775 | |
| 63 | SNP 1 | p53359-rs112371713 | Intron 5 | A, 0.1243 | AA/GA/GG | 9/160/549 | 0.4193 | 0.1651 | **0.0394** | 10/161/554 | 0.8266 | 0.2605 | 0.3391 |
| 63 | SNP 2 | p53372-rs115604379 | Intron 5 | T, 0.0066 | CC/CT | 729/10 | 3.0372 | 0.0093 | | 735/10 | 4.3130 | 0.1386 | |
| 63 | SNP 3 | p53790-rs4765614 | Intron 5 | A, 0.2653 | GG/GA/AA | 399/276/58 | -0.3281 | 0.1218 | | 402/277/59 | -0.4621 | 0.3775 | |
| 63 | SNP 4 | p54445-rs60910935 | Intron 5 | G, 0.0418 | AA/AG/GG | 650/56/2 | -0.1247 | 0.7963 | | 656/57/2 | -0.9880 | 0.4074 | |
| 64 | SNP 1 | p53372-rs115604379 | Intron 5 | T, 0.0066 | CC/CT | 729/10 | 3.0372 | 0.0093 | 0.0691 | 735/10 | 4.3130 | 0.1386 | 0.5878 |
| 64 | SNP 2 | p53790-rs4765614 | Intron 5 | A, 0.2653 | GG/GA/AA | 399/276/58 | -0.3281 | 0.1218 | | 402/277/59 | -0.4621 | 0.3775 | |
| 64 | SNP 3 | p54445-rs60910935 | Intron 5 | G, 0.0418 | AA/AG/GG | 650/56/2 | -0.1247 | 0.7963 | | 656/57/2 | -0.9880 | 0.4074 | |
| 64 | SNP 4 | p54475-rs60227139 | Intron 5 | T, 0.0437 | CC/CT/TT | 670/61/2 | -0.0602 | 0.8966 | | 676/61/2 | -0.7550 | 0.5114 | |
| 65 | SNP 1 | p53790-rs4765614 | Intron 5 | A, 0.2653 | GG/GA/AA | 399/276/58 | -0.3281 | 0.1218 | 0.4454 | 402/277/59 | -0.4621 | 0.3775 | 0.6493 |
| 65 | SNP 2 | p54445-rs60910935 | Intron 5 | G, 0.0418 | AA/AG/GG | 650/56/2 | -0.1247 | 0.7963 | | 656/57/2 | -0.9880 | 0.4074 | |
| 65 | SNP 3 | p54475-rs60227139 | Intron 5 | T, 0.0437 | CC/CT/TT | 670/61/2 | -0.0602 | 0.8966 | | 676/61/2 | -0.7550 | 0.5114 | |
| 65 | SNP 4 | p54492-rs61762481 | Intron 5 | A, 0.1005 | AA/GA/GG | 8/137/607 | -0.1522 | 0.6257 | | 8/137/618 | -0.1546 | 0.8416 | |
| 66 | SNP 1 | p54445-rs60910935 | Intron 5 | G, 0.0418 | AA/AG/GG | 650/56/2 | -0.1247 | 0.7963 | 0.4715 | 656/57/2 | -0.9880 | 0.4074 | 0.3202 |
| 66 | SNP 2 | p54475-rs60227139 | Intron 5 | T, 0.0437 | CC/CT/TT | 670/61/2 | -0.0602 | 0.8966 | | 676/61/2 | -0.7550 | 0.5114 | |
| 66 | SNP 3 | p54492-rs61762481 | Intron 5 | A, 0.1005 | AA/GA/GG | 8/137/607 | -0.1522 | 0.6257 | | 8/137/618 | -0.1546 | 0.8416 | |
| 66 | SNP 4 | p54611-chr12_125294909 | Intron 5 | C, 0.0007 | TC/TT | 1/742 | -9.5243 | 0.0097 | | 1/748 | -19.2831 | 0.0344 | |
| 67 | SNP 1 | p54475-rs60227139 | Intron 5 | T, 0.0437 | CC/CT/TT | 670/61/2 | -0.0602 | 0.8966 | 0.8784 | 676/61/2 | -0.7550 | 0.5114 | 0.8769 |
| 67 | SNP 2 | p54492-rs61762481 | Intron 5 | A, 0.1005 | AA/GA/GG | 8/137/607 | -0.1522 | 0.6257 | | 8/137/618 | -0.1546 | 0.8416 | |
| 67 | SNP 3 | p54611-chr12_125294909 | Intron 5 | C, 0.0007 | TC/TT | 1/742 | -9.5243 | 0.0097 | | 1/748 | -19.2831 | 0.0344 | |
| 67 | SNP 4 | p54627-chr12_125294893 | Intron 5 | C, 0.0020 | GC/GG | 3/733 | 0.9473 | 0.6571 | | 3/744 | 3.6910 | 0.4850 | |
| 68 | SNP 1 | p54492-rs61762481 | Intron 5 | A, 0.1005 | AA/GA/GG | 8/137/607 | -0.1522 | 0.6257 | 0.1570 | 8/137/618 | -0.1546 | 0.8416 | 0.2715 |
| 68 | SNP 2 | p54611-chr12_125294909 | Intron 5 | C, 0.0007 | TC/TT | 1/742 | -9.5243 | 0.0097 | | 1/748 | -19.2831 | 0.0344 | |
| 68 | SNP 3 | p54627-chr12_125294893 | Intron 5 | C, 0.0020 | GC/GG | 3/733 | 0.9473 | 0.6571 | | 3/744 | 3.6910 | 0.4850 | |
| 68 | SNP 4 | p54856-chr12_125294664 | Intron 6 | T, 0.0007 | CC/CT | 742/1 | -8.4305 | 0.0215 | | 748/1 | -24.0757 | 0.0082 | |
| 69 | SNP 1 | p54611-chr12_125294909 | Intron 5 | C, 0.0007 | TC/TT | 1/742 | -9.5243 | 0.0097 | 0.0610 | 1/748 | -19.2831 | 0.0344 | 0.2063 |
| 69 | SNP 2 | p54627-chr12_125294893 | Intron 5 | C, 0.0020 | GC/GG | 3/733 | 0.9473 | 0.6571 | | 3/744 | 3.6910 | 0.4850 | |
| 69 | SNP 3 | p54856-chr12_125294664 | Intron 6 | T, 0.0007 | CC/CT | 742/1 | -8.4305 | 0.0215 | | 748/1 | -24.0757 | 0.0082 | |
| 69 | SNP 4 | p55923-rs838900 | Intron 6 | A, 0.3921 | AA/GA/GG | 113/345/275 | 0.2787 | 0.1549 | | 115/346/282 | 0.3606 | 0.4549 | |
| 70 | SNP 1 | p54627-chr12_125294893 | Intron 5 | C, 0.0020 | GC/GG | 3/733 | 0.9473 | 0.6571 | 0.0599 | 3/744 | 3.6910 | 0.4850 | **0.0140** |
| 70 | SNP 2 | p54856-chr12_125294664 | Intron 6 | T, 0.0007 | CC/CT | 742/1 | -8.4305 | 0.0215 | | 748/1 | -24.0757 | 0.0082 | |
| 70 | SNP 3 | p55923-rs838900 | Intron 6 | A, 0.3921 | AA/GA/GG | 113/345/275 | 0.2787 | 0.1549 | | 115/346/282 | 0.3606 | 0.4549 | |
| 70 | SNP 4 | p55963-rs7134858 | Intron 6 | T, 0.1560 | TT/CT/CC | 24/184/532 | 0.4418 | 0.0799 | | 24/190/537 | 1.7537 | 0.0052 | |
| 71 | SNP 1 | p54856-chr12_125294664 | Intron 6 | T, 0.0007 | CC/CT | 742/1 | -8.4305 | 0.0215 | 0.0995 | 748/1 | -24.0757 | 0.0082 | **0.0488** |
| 71 | SNP 2 | p55923-rs838900 | Intron 6 | A, 0.3921 | AA/GA/GG | 113/345/275 | 0.2787 | 0.1549 | | 115/346/282 | 0.3606 | 0.4549 | |
| 71 | SNP 3 | p55963-rs7134858 | Intron 6 | T, 0.1560 | TT/CT/CC | 24/184/532 | 0.4418 | 0.0799 | | 24/190/537 | 1.7537 | 0.0052 | |
| 71 | SNP 4 | p56845-rs838902 | Intron 6 | G, 0.4249 | AA/AG/GG | 249/347/141 | -0.0786 | 0.6801 | | 256/350/143 | -0.3052 | 0.5129 | |
| 72 | SNP 1 | p55923-rs838900 | Intron 6 | A, 0.3921 | AA/GA/GG | 113/345/275 | 0.2787 | 0.1549 | **0.0315** | 115/346/282 | 0.3606 | 0.4549 | **0.0463** |
| 72 | SNP 2 | p55963-rs7134858 | Intron 6 | T, 0.1560 | TT/CT/CC | 24/184/532 | 0.4418 | 0.0799 | | 24/190/537 | 1.7537 | 0.0052 | |
| 72 | SNP 3 | p56845-rs838902 | Intron 6 | G, 0.4249 | AA/AG/GG | 249/347/141 | -0.0786 | 0.6801 | | 256/350/143 | -0.3052 | 0.5129 | |
| 72 | SNP 4 | p57004-rs187562853 | Intron 6 | A, 0.0098 | GG/GA | 721/15 | 1.6474 | 0.0872 | | 731/15 | 3.2853 | 0.1690 | |
| 73 | SNP 1 | p55963-rs7134858 | Intron 6 | T, 0.1560 | TT/CT/CC | 24/184/532 | 0.4418 | 0.0799 | 0.2741 | 24/190/537 | 1.7537 | 0.0052 | 0.0553 |
| 73 | SNP 2 | p56845-rs838902 | Intron 6 | G, 0.4249 | AA/AG/GG | 249/347/141 | -0.0786 | 0.6801 | | 256/350/143 | -0.3052 | 0.5129 | |
| 73 | SNP 3 | p57004-rs187562853 | Intron 6 | A, 0.0098 | GG/GA | 721/15 | 1.6474 | 0.0872 | | 731/15 | 3.2853 | 0.1690 | |
| 73 | SNP 4 | p57107-rs5892 | Exon 7 | T, 0.0589 | CC/CT/TT | 656/85/1 | 0.0079 | 0.9848 | | 663/84/1 | -0.4697 | 0.6488 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 74 | SNP 1 | p56845-rs838902 | Intron 6 | G, 0.4249 | AA/AG/GG | 249/347/141 | -0.0786 | 0.6801 | 0.4787 | 256/350/143 | -0.3052 | 0.5129 | 0.5963 |
| 74 | SNP 2 | p57004-rs187562853 | Intron 6 | A, 0.0098 | GG/GA | 721/15 | 1.6474 | 0.0872 | | 731/15 | 3.2853 | 0.1690 | |
| 74 | SNP 3 | p57107-rs5892 | Exon 7 | T, 0.0589 | CC/CT/TT | 656/85/1 | 0.0079 | 0.9848 | | 663/84/1 | -0.4697 | 0.6488 | |
| 74 | SNP 4 | p57508-rs71458866 | Intron 7 | A, 0.1130 | AA/GA/GG | 13/144/594 | -0.1926 | 0.5089 | | 12/147/602 | -0.6325 | 0.3861 | |
| 75 | SNP 1 | p57004-rs187562853 | Intron 6 | A, 0.0098 | GG/GA | 721/15 | 1.6474 | 0.0872 | 0.5892 | 731/15 | 3.2853 | 0.1690 | 0.2923 |
| 75 | SNP 2 | p57107-rs5892 | Exon 7 | T, 0.0589 | CC/CT/TT | 656/85/1 | 0.0079 | 0.9848 | | 663/84/1 | -0.4697 | 0.6488 | |
| 75 | SNP 3 | p57508-rs71458866 | Intron 7 | A, 0.1130 | AA/GA/GG | 13/144/594 | -0.1926 | 0.5089 | | 12/147/602 | -0.6325 | 0.3861 | |
| 75 | SNP 4 | p57592-rs838903 | Intron 7 | A, 0.3763 | AA/GA/GG | 111/334/290 | -0.1598 | 0.4149 | | 113/336/297 | -0.7661 | 0.1109 | |
| 76 | SNP 1 | p57107-rs5892 | Exon 7 | T, 0.0589 | CC/CT/TT | 656/85/1 | 0.0079 | 0.9848 | 0.9374 | 663/84/1 | -0.4697 | 0.6488 | 0.5355 |
| 76 | SNP 2 | p57508-rs71458866 | Intron 7 | A, 0.1130 | AA/GA/GG | 13/144/594 | -0.1926 | 0.5089 | | 12/147/602 | -0.6325 | 0.3861 | |
| 76 | SNP 3 | p57592-rs838903 | Intron 7 | A, 0.3763 | AA/GA/GG | 111/334/290 | -0.1598 | 0.4149 | | 113/336/297 | -0.7661 | 0.1109 | |
| 76 | SNP 4 | p58514-rs838905 | Intron 7 | C, 0.4329 | CC/TC/TT | 146/350/243 | -0.0855 | 0.6536 | | 149/352/250 | -0.4213 | 0.3646 | |
| 77 | SNP 1 | p57508-rs71458866 | Intron 7 | A, 0.1130 | AA/GA/GG | 13/144/594 | -0.1926 | 0.5089 | 0.8016 | 12/147/602 | -0.6325 | 0.3861 | 0.5014 |
| 77 | SNP 2 | p57592-rs838903 | Intron 7 | A, 0.3763 | AA/GA/GG | 111/334/290 | -0.1598 | 0.4149 | | 113/336/297 | -0.7661 | 0.1109 | |
| 77 | SNP 3 | p58514-rs838905 | Intron 7 | C, 0.4329 | CC/TC/TT | 146/350/243 | -0.0855 | 0.6536 | | 149/352/250 | -0.4213 | 0.3646 | |
| 77 | SNP 4 | p58664-rs865716 | Intron 7 | T, 0.2708 | AA/AT/TT | 395/268/62 | -0.0965 | 0.6449 | | 395/272/63 | 0.5369 | 0.3008 | |
| 78 | SNP 1 | p57592-rs838903 | Intron 7 | A, 0.3763 | AA/GA/GG | 111/334/290 | -0.1598 | 0.4149 | 0.1155 | 113/336/297 | -0.7661 | 0.1109 | **0.0326** |
| 78 | SNP 2 | p58514-rs838905 | Intron 7 | C, 0.4329 | CC/TC/TT | 146/350/243 | -0.0855 | 0.6536 | | 149/352/250 | -0.4213 | 0.3646 | |
| 78 | SNP 3 | p58664-rs865716 | Intron 7 | T, 0.2708 | AA/AT/TT | 395/268/62 | -0.0965 | 0.6449 | | 395/272/63 | 0.5369 | 0.3008 | |
| 78 | SNP 4 | p60255-rs3782287 | Intron 7 | T, 0.2831 | CC/CT/TT | 378/312/51 | 0.2555 | 0.2453 | | 384/314/55 | 0.3715 | 0.4856 | |
| 79 | SNP 1 | p58514-rs838905 | Intron 7 | C, 0.4329 | CC/TC/TT | 146/350/243 | -0.0855 | 0.6536 | 0.0820 | 149/352/250 | -0.4213 | 0.3646 | **0.0256** |
| 79 | SNP 2 | p58664-rs865716 | Intron 7 | T, 0.2708 | AA/AT/TT | 395/268/62 | -0.0965 | 0.6449 | | 395/272/63 | 0.5369 | 0.3008 | |
| 79 | SNP 3 | p60255-rs3782287 | Intron 7 | T, 0.2831 | CC/CT/TT | 378/312/51 | 0.2555 | 0.2453 | | 384/314/55 | 0.3715 | 0.4856 | |
| 79 | SNP 4 | p61872-rs838909 | Intron 7 | T, 0.2199 | CC/CT/TT | 449/249/36 | 0.1223 | 0.5945 | | 453/251/36 | 0.9232 | 0.1056 | |
| 80 | SNP 1 | p58664-rs865716 | Intron 7 | T, 0.2708 | AA/AT/TT | 395/268/62 | -0.0965 | 0.6449 | 0.1146 | 395/272/63 | 0.5369 | 0.3008 | **0.0030** |
| 80 | SNP 2 | p60255-rs3782287 | Intron 7 | T, 0.2831 | CC/CT/TT | 378/312/51 | 0.2555 | 0.2453 | | 384/314/55 | 0.3715 | 0.4856 | |
| 80 | SNP 3 | p61872-rs838909 | Intron 7 | T, 0.2199 | CC/CT/TT | 449/249/36 | 0.1223 | 0.5945 | | 453/251/36 | 0.9232 | 0.1056 | |
| 80 | SNP 4 | p62140-rs838910 | Intron 7 | T, 0.3047 | GG/GT/TT | 355/304/70 | -0.0759 | 0.7143 | | 358/304/72 | -0.0755 | 0.8821 | |
| 81 | SNP 1 | p60255-rs3782287 | Intron 7 | T, 0.2831 | CC/CT/TT | 378/312/51 | 0.2555 | 0.2453 | 0.2078 | 384/314/55 | 0.3715 | 0.4856 | **0.0050** |
| 81 | SNP 2 | p61872-rs838909 | Intron 7 | T, 0.2199 | CC/CT/TT | 449/249/36 | 0.1223 | 0.5945 | | 453/251/36 | 0.9232 | 0.1056 | |
| 81 | SNP 3 | p62140-rs838910 | Intron 7 | T, 0.3047 | GG/GT/TT | 355/304/70 | -0.0759 | 0.7143 | | 358/304/72 | -0.0755 | 0.8821 | |
| 81 | SNP 4 | p62409-rs838911 | Intron 7 | T, 0.4211 | CC/CT/TT | 247/347/133 | -0.1556 | 0.4213 | | 251/346/135 | -0.6245 | 0.1888 | |
| 82 | SNP 1 | p61872-rs838909 | Intron 7 | T, 0.2199 | CC/CT/TT | 449/249/36 | 0.1223 | 0.5945 | 0.1998 | 453/251/36 | 0.9232 | 0.1056 | **0.0137** |
| 82 | SNP 2 | p62140-rs838910 | Intron 7 | T, 0.3047 | GG/GT/TT | 355/304/70 | -0.0759 | 0.7143 | | 358/304/72 | -0.0755 | 0.8821 | |
| 82 | SNP 3 | p62409-rs838911 | Intron 7 | T, 0.4211 | CC/CT/TT | 247/347/133 | -0.1556 | 0.4213 | | 251/346/135 | -0.6245 | 0.1888 | |
| 82 | SNP 4 | p62615-rs7138386 | Intron 7 | C, 0.1137 | TT/TC/CC | 568/138/12 | -0.2083 | 0.4851 | | 571/141/11 | -0.6495 | 0.3851 | |
| 83 | SNP 1 | p62140-rs838910 | Intron 7 | T, 0.3047 | GG/GT/TT | 355/304/70 | -0.0759 | 0.7143 | 0.2052 | 358/304/72 | -0.0755 | 0.8821 | **0.0187** |
| 83 | SNP 2 | p62409-rs838911 | Intron 7 | T, 0.4211 | CC/CT/TT | 247/347/133 | -0.1556 | 0.4213 | | 251/346/135 | -0.6245 | 0.1888 | |
| 83 | SNP 3 | p62615-rs7138386 | Intron 7 | C, 0.1137 | TT/TC/CC | 568/138/12 | -0.2083 | 0.4851 | | 571/141/11 | -0.6495 | 0.3851 | |
| 83 | SNP 4 | p63483-rs838912 | Intron 7 | A, 0.0867 | AA/GA/GG | 6/117/615 | 0.1814 | 0.5840 | | 6/119/624 | 1.8700 | 0.0234 | |
| 84 | SNP 1 | p62409-rs838911 | Intron 7 | T, 0.4211 | CC/CT/TT | 247/347/133 | -0.1556 | 0.4213 | 0.7674 | 251/346/135 | -0.6245 | 0.1888 | 0.1544 |
| 84 | SNP 2 | p62615-rs7138386 | Intron 7 | C, 0.1137 | TT/TC/CC | 568/138/12 | -0.2083 | 0.4851 | | 571/141/11 | -0.6495 | 0.3851 | |
| 84 | SNP 3 | p63483-rs838912 | Intron 7 | A, 0.0867 | AA/GA/GG | 6/117/615 | 0.1814 | 0.5840 | | 6/119/624 | 1.8700 | 0.0234 | |
| 84 | SNP 4 | p64772-rs5888 | Exon 8 | T, 0.0961 | CC/CT/TT | 605/129/8 | 0.3620 | 0.2561 | | 614/130/8 | 2.0962 | 0.0080 | |
| 85 | SNP 1 | p62615-rs7138386 | Intron 7 | C, 0.1137 | TT/TC/CC | 568/138/12 | -0.2083 | 0.4851 | 0.7618 | 571/141/11 | -0.6495 | 0.3851 | 0.1130 |
| 85 | SNP 2 | p63483-rs838912 | Intron 7 | A, 0.0867 | AA/GA/GG | 6/117/615 | 0.1814 | 0.5840 | | 6/119/624 | 1.8700 | 0.0234 | |
| 85 | SNP 3 | p64772-rs5888 | Exon 8 | T, 0.0961 | CC/CT/TT | 605/129/8 | 0.3620 | 0.2561 | | 614/130/8 | 2.0962 | 0.0080 | |
| 85 | SNP 4 | p64923-rs838915 | Intron 8 | A, 0.1435 | AA/CA/CC | 19/177/539 | -0.0858 | 0.7466 | | 19/179/547 | -0.3684 | 0.5766 | |
| 86 | SNP 1 | p63483-rs838912 | Intron 7 | A, 0.0867 | AA/GA/GG | 6/117/615 | 0.1814 | 0.5840 | 0.1859 | 6/119/624 | 1.8700 | 0.0234 | **0.0290** |
| 86 | SNP 2 | p64772-rs5888 | Exon 8 | T, 0.0961 | CC/CT/TT | 605/129/8 | 0.3620 | 0.2561 | | 614/130/8 | 2.0962 | 0.0080 | |
| 86 | SNP 3 | p64923-rs838915 | Intron 8 | A, 0.1435 | AA/CA/CC | 19/177/539 | -0.0858 | 0.7466 | | 19/179/547 | -0.3684 | 0.5766 | |
| 86 | SNP 4 | p65999-rs12819677 | Intron 8 | A, 0.2813 | GG/GA/AA | 371/310/50 | 0.4021 | 0.0670 | | 375/309/52 | 0.6769 | 0.2052 | |
| 87 | SNP 1 | p64772-rs5888 | Exon 8 | T, 0.0961 | CC/CT/TT | 605/129/8 | 0.3620 | 0.2561 | 0.1207 | 614/130/8 | 2.0962 | 0.0080 | 0.0623 |
| 87 | SNP 2 | p64923-rs838915 | Intron 8 | A, 0.1435 | AA/CA/CC | 19/177/539 | -0.0858 | 0.7466 | | 19/179/547 | -0.3684 | 0.5766 | |
| 87 | SNP 3 | p65999-rs12819677 | Intron 8 | A, 0.2813 | GG/GA/AA | 371/310/50 | 0.4021 | 0.0670 | | 375/309/52 | 0.6769 | 0.2052 | |
| 87 | SNP 4 | p67439-rs961170 | Intron 8 | A, 0.0893 | AA/GA/GG | 11/108/610 | -0.0481 | 0.8812 | | 11/108/615 | -0.9808 | 0.2211 | |
| 88 | SNP 1 | p64923-rs838915 | Intron 8 | A, 0.1435 | AA/CA/CC | 19/177/539 | -0.0858 | 0.7466 | 0.2674 | 19/179/547 | -0.3684 | 0.5766 | 0.1964 |
| 88 | SNP 2 | p65999-rs12819677 | Intron 8 | A, 0.2813 | GG/GA/AA | 371/310/50 | 0.4021 | 0.0670 | | 375/309/52 | 0.6769 | 0.2052 | |
| 88 | SNP 3 | p67439-rs961170 | Intron 8 | A, 0.0893 | AA/GA/GG | 11/108/610 | -0.0481 | 0.8812 | | 11/108/615 | -0.9808 | 0.2211 | |
| 88 | SNP 4 | p67700-rs1726374 | Intron 8 | A, 0.1933 | GG/GA/AA | 491/221/35 | 0.2289 | 0.3262 | | 500/226/32 | 0.3249 | 0.5798 | |
| 89 | SNP 1 | p65999-rs12819677 | Intron 8 | A, 0.2813 | GG/GA/AA | 371/310/50 | 0.4021 | 0.0670 | 0.2789 | 375/309/52 | 0.6769 | 0.2052 | 0.1543 |
| 89 | SNP 2 | p67439-rs961170 | Intron 8 | A, 0.0893 | AA/GA/GG | 11/108/610 | -0.0481 | 0.8812 | | 11/108/615 | -0.9808 | 0.2211 | |
| 89 | SNP 3 | p67700-rs1726374 | Intron 8 | A, 0.1933 | GG/GA/AA | 491/221/35 | 0.2289 | 0.3262 | | 500/226/32 | 0.3249 | 0.5798 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 89 | SNP 4 | p69013-rs7135117 | Intron 8 | G, 0.2901 | GG/AG/AA | 75/275/377 | 0.1133 | 0.5758 | | 74/275/383 | 0.5010 | 0.3196 | |
| 90 | SNP 1 | p67439-rs961170 | Intron 8 | A, 0.0893 | AA/GA/GG | 11/108/610 | -0.0481 | 0.8812 | 0.4209 | 11/108/615 | -0.9808 | 0.2211 | 0.1317 |
| 90 | SNP 2 | p67700-rs1726374 | Intron 8 | A, 0.1933 | GG/GA/AA | 491/221/35 | 0.2289 | 0.3262 | | 500/226/32 | 0.3249 | 0.5798 | |
| 90 | SNP 3 | p69013-rs7135117 | Intron 8 | G, 0.2901 | GG/AG/AA | 75/275/377 | 0.1133 | 0.5758 | | 74/275/383 | 0.5010 | 0.3196 | |
| 90 | SNP 4 | p69699-rs10396210 | Intron 8-splice site | A, 0.1511 | AA/GA/GG | 17/186/523 | -0.4778 | 0.0745 | | 17/188/527 | -1.2921 | 0.0520 | |
| 91 | SNP 1 | p67700-rs1726374 | Intron 8 | A, 0.1933 | GG/GA/AA | 491/221/35 | 0.2289 | 0.3262 | 0.4715 | 500/226/32 | 0.3249 | 0.5798 | 0.4607 |
| 91 | SNP 2 | p69013-rs7135117 | Intron 8 | G, 0.2901 | GG/AG/AA | 75/275/377 | 0.1133 | 0.5758 | | 74/275/383 | 0.5010 | 0.3196 | |
| 91 | SNP 3 | p69699-rs10396210 | Intron 8-splice site | A, 0.1511 | AA/GA/GG | 17/186/523 | -0.4778 | 0.0745 | | 17/188/527 | -1.2921 | 0.0520 | |
| 91 | SNP 4 | p69995delC-rs5801571 | Intron 9 | delC, 0.2761 | DD/WD/WW | 63/281/386 | 0.0614 | 0.7724 | | 63/282/394 | 0.2920 | 0.5768 | |
| 92 | SNP 1 | p69013-rs7135117 | Intron 8 | G, 0.2901 | GG/AG/AA | 75/275/377 | 0.1133 | 0.5758 | 0.2848 | 74/275/383 | 0.5010 | 0.3196 | 0.4449 |
| 92 | SNP 2 | p69699-rs10396210 | Intron 8-splice site | A, 0.1511 | AA/GA/GG | 17/186/523 | -0.4778 | 0.0745 | | 17/188/527 | -1.2921 | 0.0520 | |
| 92 | SNP 3 | p69995delC-rs5801571 | Intron 9 | delC, 0.2761 | DD/WD/WW | 63/281/386 | 0.0614 | 0.7724 | | 63/282/394 | 0.2920 | 0.5768 | |
| 92 | SNP 4 | p71867-rs7954022 | Intron 9 | T, 0.1323 | TT/CT/CC | 11/172/552 | 0.3876 | 0.1764 | | 12/172/556 | 0.8502 | 0.2241 | |
| 93 | SNP 1 | p69699-rs10396210 | Intron 8-splice site | A, 0.1511 | AA/GA/GG | 17/186/523 | -0.4778 | 0.0745 | 0.6336 | 17/188/527 | -1.2921 | 0.0520 | 0.2477 |
| 93 | SNP 2 | p69995delC-rs5801571 | Intron 9 | delC, 0.2761 | DD/WD/WW | 63/281/386 | 0.0614 | 0.7724 | | 63/282/394 | 0.2920 | 0.5768 | |
| 93 | SNP 3 | p71867-rs7954022 | Intron 9 | T, 0.1323 | TT/CT/CC | 11/172/552 | 0.3876 | 0.1764 | | 12/172/556 | 0.8502 | 0.2241 | |
| 93 | SNP 4 | p72197-rs838861 | Intron 9 | G, 0.3777 | AA/AG/GG | 300/308/123 | -0.1415 | 0.4527 | | 304/310/122 | -0.1507 | 0.7464 | |
| 94 | SNP 1 | p69995delC-rs5801571 | Intron 9 | delC, 0.2761 | DD/WD/WW | 63/281/386 | 0.0614 | 0.7724 | 0.7410 | 63/282/394 | 0.2920 | 0.5768 | 0.3380 |
| 94 | SNP 2 | p71867-rs7954022 | Intron 9 | T, 0.1323 | TT/CT/CC | 11/172/552 | 0.3876 | 0.1764 | | 12/172/556 | 0.8502 | 0.2241 | |
| 94 | SNP 3 | p72197-rs838861 | Intron 9 | G, 0.3777 | AA/AG/GG | 300/308/123 | -0.1415 | 0.4527 | | 304/310/122 | -0.1507 | 0.7464 | |
| 94 | SNP 4 | p72777-rs838862 | Intron 9 | T, 0.0887 | CC/CT/TT | 607/115/7 | 0.1013 | 0.7613 | | 611/116/7 | 0.7012 | 0.3938 | |
| 95 | SNP 1 | p71867-rs7954022 | Intron 9 | T, 0.1323 | TT/CT/CC | 11/172/552 | 0.3876 | 0.1764 | 0.1054 | 12/172/556 | 0.8502 | 0.2241 | **0.0131** |
| 95 | SNP 2 | p72197-rs838861 | Intron 9 | G, 0.3777 | AA/AG/GG | 300/308/123 | -0.1415 | 0.4527 | | 304/310/122 | -0.1507 | 0.7464 | |
| 95 | SNP 3 | p72777-rs838862 | Intron 9 | T, 0.0887 | CC/CT/TT | 607/115/7 | 0.1013 | 0.7613 | | 611/116/7 | 0.7012 | 0.3938 | |
| 95 | SNP 4 | p75766-rs838866 | Intron 9 | C, 0.2116 | TT/TC/CC | 457/239/38 | -0.1058 | 0.6423 | | 466/240/37 | -0.0497 | 0.9306 | |
| 96 | SNP 1 | p72197-rs838861 | Intron 9 | G, 0.3777 | AA/AG/GG | 300/308/123 | -0.1415 | 0.4527 | 0.4137 | 304/310/122 | -0.1507 | 0.7464 | **0.0484** |
| 96 | SNP 2 | p72777-rs838862 | Intron 9 | T, 0.0887 | CC/CT/TT | 607/115/7 | 0.1013 | 0.7613 | | 611/116/7 | 0.7012 | 0.3938 | |
| 96 | SNP 3 | p75766-rs838866 | Intron 9 | C, 0.2116 | TT/TC/CC | 457/239/38 | -0.1058 | 0.6423 | | 466/240/37 | -0.0497 | 0.9306 | |
| 96 | SNP 4 | p75778-rs7301120 | Intron 9 | T, 0.1135 | TT/CT/CC | 9/147/563 | -0.2367 | 0.4366 | | 9/146/569 | 0.3767 | 0.6174 | |
| 97 | SNP 1 | p72777-rs838862 | Intron 9 | T, 0.0887 | CC/CT/TT | 607/115/7 | 0.1013 | 0.7613 | 0.0867 | 611/116/7 | 0.7012 | 0.3938 | **0.0098** |
| 97 | SNP 2 | p75766-rs838866 | Intron 9 | C, 0.2116 | TT/TC/CC | 457/239/38 | -0.1058 | 0.6423 | | 466/240/37 | -0.0497 | 0.9306 | |
| 97 | SNP 3 | p75778-rs7301120 | Intron 9 | T, 0.1135 | TT/CT/CC | 9/147/563 | -0.2367 | 0.4366 | | 9/146/569 | 0.3767 | 0.6174 | |
| 97 | SNP 4 | p76757-rs9919713 | Intron 9 | T, 0.4390 | AA/AT/TT | 235/347/151 | -0.1264 | 0.5044 | | 243/353/148 | -0.1860 | 0.6921 | |
| 98 | SNP 1 | p75766-rs838866 | Intron 9 | C, 0.2116 | TT/TC/CC | 457/239/38 | -0.1058 | 0.6423 | 0.5187 | 466/240/37 | -0.0497 | 0.9306 | 0.7976 |
| 98 | SNP 2 | p75778-rs7301120 | Intron 9 | T, 0.1135 | TT/CT/CC | 9/147/563 | -0.2367 | 0.4366 | | 9/146/569 | 0.3767 | 0.6174 | |
| 98 | SNP 3 | p76757-rs9919713 | Intron 9 | T, 0.4390 | AA/AT/TT | 235/347/151 | -0.1264 | 0.5044 | | 243/353/148 | -0.1860 | 0.6921 | |
| 98 | SNP 4 | p77181-rs146246031 | Intron 9 | C, 0.0053 | TC/TT | 6/731 | -0.6173 | 0.6809 | | 8/734 | 0.4564 | 0.8874 | |
| 99 | SNP 1 | p75778-rs7301120 | Intron 9 | T, 0.1135 | TT/CT/CC | 9/147/563 | -0.2367 | 0.4366 | 0.7604 | 9/146/569 | 0.3767 | 0.6174 | 0.5753 |
| 99 | SNP 2 | p76757-rs9919713 | Intron 9 | T, 0.4390 | AA/AT/TT | 235/347/151 | -0.1264 | 0.5044 | | 243/353/148 | -0.1860 | 0.6921 | |
| 99 | SNP 3 | p77181-rs146246031 | Intron 9 | C, 0.0053 | TC/TT | 6/731 | -0.6173 | 0.6809 | | 8/734 | 0.4564 | 0.8874 | |
| 99 | SNP 4 | p77251-rs34339961 | Intron 9 | T, 0.1177 | AA/AT/TT | 561/151/11 | -0.1372 | 0.6426 | | 573/150/11 | 0.7120 | 0.3383 | |
| 100 | SNP 1 | p76757-rs9919713 | Intron 9 | T, 0.439 | AA/AT/TT | 235/347/151 | -0.1264 | 0.5044 | 0.8818 | 243/353/148 | -0.1860 | 0.6921 | 0.6540 |
| 100 | SNP 2 | p77181-rs146246031 | Intron 9 | C, 0.0053 | TC/TT | 6/731 | -0.6173 | 0.6809 | | 8/734 | 0.4564 | 0.8874 | |
| 100 | SNP 3 | p77251-rs34339961 | Intron 9 | T, 0.1177 | AA/AT/TT | 561/151/11 | -0.1372 | 0.6426 | | 573/150/11 | 0.7120 | 0.3383 | |
| 100 | SNP 4 | p77381-rs138499966 | Intron 9 | C, 0.0046 | TC/TT | 7/735 | 1.5523 | 0.2652 | | 7/741 | -0.2236 | 0.9485 | |
| 101 | SNP 1 | p77181-rs146246031 | Intron 9 | C, 0.0053 | TC/TT | 6/731 | -0.6173 | 0.6809 | 0.7886 | 8/734 | 0.4564 | 0.8874 | 0.6293 |
| 101 | SNP 2 | p77251-rs34339961 | Intron 9 | T, 0.1177 | AA/AT/TT | 561/151/11 | -0.1372 | 0.6426 | | 573/150/11 | 0.7120 | 0.3383 | |
| 101 | SNP 3 | p77381-rs138499966 | Intron 9 | C, 0.0046 | TC/TT | 7/735 | 1.5523 | 0.2652 | | 7/741 | -0.2236 | 0.9485 | |
| 101 | SNP 4 | p77620-rs377124254 | Intron 10 | A, 0.0007 | GA/GG | 1/735 | 11.5518 | 0.0016 | | 1/741 | 14.4685 | 0.1141 | |
| 102 | SNP 1 | p77251-rs34339961 | Intron 9 | T, 0.1177 | AA/AT/TT | 561/151/11 | -0.1372 | 0.6426 | 0.5420 | 573/150/11 | 0.7120 | 0.3383 | 0.4369 |
| 102 | SNP 2 | p77381-rs138499966 | Intron 9 | C, 0.0046 | TC/TT | 7/735 | 1.5523 | 0.2652 | | 7/741 | -0.2236 | 0.9485 | |
| 102 | SNP 3 | p77620-rs377124254 | Intron 10 | A, 0.0007 | GA/GG | 1/735 | 11.5518 | 0.0016 | | 1/741 | 14.4685 | 0.1141 | |
| 102 | SNP 4 | p77682-rs150082885 | Intron 10 | G, 0.0106 | AA/AG/GG | 716/13/1 | -0.5377 | 0.5509 | | 721/14/1 | -2.9877 | 0.1698 | |
| 103 | SNP 1 | p77381-rs138499966 | Intron 9 | C, 0.0046 | TC/TT | 7/735 | 1.5523 | 0.2652 | 0.8039 | 7/741 | -0.2236 | 0.9485 | 0.3187 |
| 103 | SNP 2 | p77620-rs377124254 | Intron 10 | A, 0.0007 | GA/GG | 1/735 | 11.5518 | 0.0016 | | 1/741 | 14.4685 | 0.1141 | |
| 103 | SNP 3 | p77682-rs150082885 | Intron 10 | G, 0.0106 | AA/AG/GG | 716/13/1 | -0.5377 | 0.5509 | | 721/14/1 | -2.9877 | 0.1698 | |
| 103 | SNP 4 | p77704-chr12_125271816 | Intron 10 | A, 0.0040 | CA/CC | 5/726 | -1.4602 | 0.3791 | | 6/731 | -3.0744 | 0.4152 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 104 | SNP 1 | p77620-rs377124254 | Intron 10 | A, 0.0007 | GA/GG | 1/735 | 11.5518 | 0.0016 | 0.7533 | 1/741 | 14.4685 | 0.1141 | 0.3571 |
| 104 | SNP 2 | p77682-rs150082885 | Intron 10 | G, 0.0106 | AA/AG/GG | 716/13/1 | -0.5377 | 0.5509 | | 721/14/1 | -2.9877 | 0.1698 | |
| 104 | SNP 3 | p77704-chr12_125271816 | Intron 10 | A, 0.0040 | CA/CC | 5/726 | -1.4602 | 0.3791 | | 6/731 | -3.0744 | 0.4152 | |
| 104 | SNP 4 | p77842-rs2272310 | Intron 10 | A, 0.0807 | AA/GA/GG | 5/110/630 | 0.2011 | 0.5651 | | 5/110/642 | 0.8339 | 0.3359 | |
| 105 | SNP 1 | p77682-rs150082885 | Intron 10 | G, 0.0106 | AA/AG/GG | 716/13/1 | -0.5377 | 0.5509 | 0.8340 | 721/14/1 | -2.9877 | 0.1698 | 0.4218 |
| 105 | SNP 2 | p77704-chr12_125271816 | Intron 10 | A, 0.0040 | CA/CC | 5/726 | -1.4602 | 0.3791 | | 6/731 | -3.0744 | 0.4152 | |
| 105 | SNP 3 | p77842-rs2272310 | Intron 10 | A, 0.0807 | AA/GA/GG | 5/110/630 | 0.2011 | 0.5651 | | 5/110/642 | 0.8339 | 0.3359 | |
| 105 | SNP 4 | p78255-rs184052375 | Intron 10 | G, 0.0072 | AA/AG | 732/11 | 1.1808 | 0.2893 | | 740/9 | 3.3097 | 0.2790 | |
| 106 | SNP 1 | p77704-chr12_125271816 | Intron 10 | A, 0.0040 | CA/CC | 5/726 | -1.4602 | 0.3791 | 0.9158 | 6/731 | -3.0744 | 0.4152 | 0.3479 |
| 106 | SNP 2 | p77842-rs2272310 | Intron 10 | A, 0.0807 | AA/GA/GG | 5/110/630 | 0.2011 | 0.5651 | | 5/110/642 | 0.8339 | 0.3359 | |
| 106 | SNP 3 | p78255-rs184052375 | Intron 10 | G, 0.0072 | AA/AG | 732/11 | 1.1808 | 0.2893 | | 740/9 | 3.3097 | 0.2790 | |
| 106 | SNP 4 | p78402-rs838898 | Intron 10 | A, 0.0714 | AA/GA/GG | 7/86/594 | -0.0602 | 0.8720 | | 7/86/601 | -0.9806 | 0.2889 | |
| 107 | SNP 1 | p77842-rs2272310 | Intron 10 | A, 0.0807 | AA/GA/GG | 5/110/630 | 0.2011 | 0.5651 | 0.8730 | 5/110/642 | 0.8339 | 0.3359 | 0.5898 |
| 107 | SNP 2 | p78255-rs184052375 | Intron 10 | G, 0.0072 | AA/AG | 732/11 | 1.1808 | 0.2893 | | 740/9 | 3.3097 | 0.2790 | |
| 107 | SNP 3 | p78402-rs838898 | Intron 10 | A, 0.0714 | AA/GA/GG | 7/86/594 | -0.0602 | 0.8720 | | 7/86/601 | -0.9806 | 0.2889 | |
| 107 | SNP 4 | p78430-rs838897 | Intron 10 | G, 0.3830 | GG/CG/CC | 125/308/291 | 0.0070 | 0.9704 | | 121/315/299 | -0.1887 | 0.6887 | |
| 108 | SNP 1 | p78255-rs184052375 | Intron 10 | G, 0.0072 | AA/AG | 732/11 | 1.1808 | 0.2893 | 0.4248 | 740/9 | 3.3097 | 0.2790 | 0.0975 |
| 108 | SNP 2 | p78402-rs838898 | Intron 10 | A, 0.0714 | AA/GA/GG | 7/86/594 | -0.0602 | 0.8720 | | 7/86/601 | -0.9806 | 0.2889 | |
| 108 | SNP 3 | p78430-rs838897 | Intron 10 | G, 0.3830 | GG/CG/CC | 125/308/291 | 0.0070 | 0.9704 | | 121/315/299 | -0.1887 | 0.6887 | |
| 108 | SNP 4 | p78747-rs2293440 | Intron 11 | C, 0.4112 | CC/TC/TT | 128/342/252 | -0.1684 | 0.3806 | | 126/348/258 | -0.2984 | 0.5352 | |
| 109 | SNP 1 | p78402-rs838898 | Intron 10 | A, 0.0714 | AA/GA/GG | 7/86/594 | -0.0602 | 0.8720 | 0.4162 | 7/86/601 | -0.9806 | 0.2889 | **0.0195** |
| 109 | SNP 2 | p78430-rs838897 | Intron 10 | G, 0.3830 | GG/CG/CC | 125/308/291 | 0.0070 | 0.9704 | | 121/315/299 | -0.1887 | 0.6887 | |
| 109 | SNP 3 | p78747-rs2293440 | Intron 11 | C, 0.4112 | CC/TC/TT | 128/342/252 | -0.1684 | 0.3806 | | 126/348/258 | -0.2984 | 0.5352 | |
| 109 | SNP 4 | p78791-rs75289200 | Intron 11 | C, 0.0321 | TC/TT | 46/679 | 0.7037 | 0.2078 | | 46/685 | 3.6568 | 0.0086 | |
| 110 | SNP 1 | p78430-rs838897 | Intron 10 | G, 0.3830 | GG/CG/CC | 125/308/291 | 0.0070 | 0.9704 | 0.1013 | 121/315/299 | -0.1887 | 0.6887 | **0.0012** |
| 110 | SNP 2 | p78747-rs2293440 | Intron 11 | C, 0.4112 | CC/TC/TT | 128/342/252 | -0.1684 | 0.3806 | | 126/348/258 | -0.2984 | 0.5352 | |
| 110 | SNP 3 | p78791-rs75289200 | Intron 11 | C, 0.0321 | TC/TT | 46/679 | 0.7037 | 0.2078 | | 46/685 | 3.6568 | 0.0086 | |
| 110 | SNP 4 | p79721-rs838896 | Intron 11 | C, 0.3104 | GG/GC/CC | 349/319/73 | 0.3565 | 0.0817 | | 357/324/71 | 1.1147 | 0.0278 | |
| 111 | SNP 1 | p78747-rs2293440 | Intron 11 | C, 0.4112 | CC/TC/TT | 128/342/252 | -0.1684 | 0.3806 | **0.0040** | 126/348/258 | -0.2984 | 0.5352 | **0.0038** |
| 111 | SNP 2 | p78791-rs75289200 | Intron 11 | C, 0.0321 | TC/TT | 46/679 | 0.7037 | 0.2078 | | 46/685 | 3.6568 | 0.0086 | |
| 111 | SNP 3 | p79721-rs838896 | Intron 11 | C, 0.3104 | GG/GC/CC | 349/319/73 | 0.3565 | 0.0817 | | 357/324/71 | 1.1147 | 0.0278 | |
| 111 | SNP 4 | p79828-rs838895 | Intron 11 | G, 0.3171 | GG/CG/CC | 74/322/337 | 0.4961 | 0.0162 | | 73/325/345 | 1.2206 | 0.0164 | |
| 112 | SNP 1 | p78791-rs75289200 | Intron 11 | C, 0.0321 | TC/TT | 46/679 | 0.7037 | 0.2078 | **0.0055** | 46/685 | 3.6568 | 0.0086 | **0.0412** |
| 112 | SNP 2 | p79721-rs838896 | Intron 11 | C, 0.3104 | GG/GC/CC | 349/319/73 | 0.3565 | 0.0817 | | 357/324/71 | 1.1147 | 0.0278 | |
| 112 | SNP 3 | p79828-rs838895 | Intron 11 | G, 0.3171 | GG/CG/CC | 74/322/337 | 0.4961 | 0.0162 | | 73/325/345 | 1.2206 | 0.0164 | |
| 112 | SNP 4 | p80045-rs838893 | Intron 11 | A, 0.3244 | GG/GA/AA | 335/325/81 | 0.3127 | 0.1224 | | 344/328/80 | 0.8859 | 0.0774 | |
| 113 | SNP 1 | p79721-rs838896 | Intron 11 | C, 0.3104 | GG/GC/CC | 349/319/73 | 0.3565 | 0.0817 | **0.0048** | 357/324/71 | 1.1147 | 0.0278 | 0.1581 |
| 113 | SNP 2 | p79828-rs838895 | Intron 11 | G, 0.3171 | GG/CG/CC | 74/322/337 | 0.4961 | 0.0162 | | 73/325/345 | 1.2206 | 0.0164 | |
| 113 | SNP 3 | p80045-rs838893 | Intron 11 | A, 0.3244 | GG/GA/AA | 335/325/81 | 0.3127 | 0.1224 | | 344/328/80 | 0.8859 | 0.0774 | |
| 113 | SNP 4 | p81863-rs185445624 | Intron 11 | A, 0.0020 | GA/GG | 3/739 | -0.9612 | 0.6510 | | 3/745 | -2.5166 | 0.6333 | |
| 114 | SNP 1 | p79828-rs838895 | Intron 11 | G, 0.3171 | GG/CG/CC | 74/322/337 | 0.4961 | 0.0162 | **0.0447** | 73/325/345 | 1.2206 | 0.0164 | 0.1200 |
| 114 | SNP 2 | p80045-rs838893 | Intron 11 | A, 0.3244 | GG/GA/AA | 335/325/81 | 0.3127 | 0.1224 | | 344/328/80 | 0.8859 | 0.0774 | |
| 114 | SNP 3 | p81863-rs185445624 | Intron 11 | A, 0.0020 | GA/GG | 3/739 | -0.9612 | 0.6510 | | 3/745 | -2.5166 | 0.6333 | |
| 114 | SNP 4 | p82019-rs838890 | Intron 11 | T, 0.0320 | CC/CT/TT | 683/42/2 | -1.0051 | 0.0618 | | 690/41/2 | -1.1864 | 0.3782 | |
| 115 | SNP 1 | p80045-rs838893 | Intron 11 | A, 0.3244 | GG/GA/AA | 335/325/81 | 0.3127 | 0.1224 | 0.0934 | 344/328/80 | 0.8859 | 0.0774 | 0.2923 |
| 115 | SNP 2 | p81863-rs185445624 | Intron 11 | A, 0.0020 | GA/GG | 3/739 | -0.9612 | 0.6510 | | 3/745 | -2.5166 | 0.6333 | |
| 115 | SNP 3 | p82019-rs838890 | Intron 11 | T, 0.0320 | CC/CT/TT | 683/42/2 | -1.0051 | 0.0618 | | 690/41/2 | -1.1864 | 0.3782 | |
| 115 | SNP 4 | p82264-rs141545424 | Exon 12 | A, 0.0007 | CA/CC | 1/739 | 11.5850 | 0.0016 | | 1/745 | 14.4986 | 0.1119 | |
| 116 | SNP 1 | p81863-rs185445624 | Intron 11 | A, 0.0020 | GA/GG | 3/739 | -0.9612 | 0.6510 | 0.0537 | 3/745 | -2.5166 | 0.6333 | 0.5297 |
| 116 | SNP 2 | p82019-rs838890 | Intron 11 | T, 0.0320 | CC/CT/TT | 683/42/2 | -1.0051 | 0.0618 | | 690/41/2 | -1.1864 | 0.3782 | |
| 116 | SNP 3 | p82264-rs141545424 | Exon 12 | A, 0.0007 | CA/CC | 1/739 | 11.5850 | 0.0016 | | 1/745 | 14.4986 | 0.1119 | |
| 116 | SNP 4 | p82340-rs77483223 | Intron 12 | A, 0.0231 | GA/GG | 35/699 | -1.0458 | 0.1012 | | 35/705 | -1.5601 | 0.3268 | |
| 117 | SNP 1 | p82019-rs838890 | Intron 11 | T, 0.0320 | CC/CT/TT | 683/42/2 | -1.0051 | 0.0618 | **0.0433** | 690/41/2 | -1.1864 | 0.3782 | 0.3386 |
| 117 | SNP 2 | p82264-rs141545424 | Exon 12 | A, 0.0007 | CA/CC | 1/739 | 11.5850 | 0.0016 | | 1/745 | 14.4986 | 0.1119 | |
| 117 | SNP 3 | p82340-rs77483223 | Intron 12 | A, 0.0231 | GA/GG | 35/699 | -1.0458 | 0.1012 | | 35/705 | -1.5601 | 0.3268 | |
| 117 | SNP 4 | p82369-rs75446635 | Intron 12 | A, 0.0059 | GA/GG | 9/733 | 0.5896 | 0.6322 | | 9/739 | 2.2239 | 0.4660 | |
| 118 | SNP 1 | p82264-rs141545424 | Exon 12 | A, 0.0007 | CA/CC | 1/739 | 11.5850 | 0.0016 | **0.0375** | 1/745 | 14.4986 | 0.1119 | 0.3863 |
| 118 | SNP 2 | p82340-rs77483223 | Intron 12 | A, 0.0231 | GA/GG | 35/699 | -1.0458 | 0.1012 | | 35/705 | -1.5601 | 0.3268 | |
| 118 | SNP 3 | p82369-rs75446635 | Intron 12 | A, 0.0059 | GA/GG | 9/733 | 0.5896 | 0.6322 | | 9/739 | 2.2239 | 0.4660 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 118 | SNP 4 | p82434-rs838889 | Intron 12 | C, 0.0315 | CC/TC/TT | 2/42/695 | -1.0389 | 0.0526 | | 2/41/702 | -0.9096 | 0.4967 | |
| 119 | SNP 1 | p82340-rs77483223 | Intron 12 | A, 0.0231 | GA/GG | 35/699 | -1.0458 | 0.1012 | 0.2138 | 35/705 | -1.5601 | 0.3268 | 0.7897 |
| 119 | SNP 2 | p82369-rs75446635 | Intron 12 | A, 0.0059 | GA/GG | 9/733 | 0.5896 | 0.6322 | | 9/739 | 2.2239 | 0.4660 | |
| 119 | SNP 3 | p82434-rs838889 | Intron 12 | C, 0.0315 | CC/TC/TT | 2/42/695 | -1.0389 | 0.0526 | | 2/41/702 | -0.9096 | 0.4967 | |
| 119 | SNP 4 | p83547-rs838887 | Intron 12 | G, 0.4564 | CC/CG/GG | 225/357/154 | 0.1202 | 0.5267 | | 227/358/157 | -0.1113 | 0.8119 | |
| 120 | SNP 1 | p82369-rs75446635 | Intron 12 | A, 0.0059 | GA/GG | 9/733 | 0.5896 | 0.6322 | 0.3753 | 9/739 | 2.2239 | 0.4660 | 0.2078 |
| 120 | SNP 2 | p82434-rs838889 | Intron 12 | C, 0.0315 | CC/TC/TT | 2/42/695 | -1.0389 | 0.0526 | | 2/41/702 | -0.9096 | 0.4967 | |
| 120 | SNP 3 | p83547-rs838887 | Intron 12 | G, 0.4564 | CC/CG/GG | 225/357/154 | 0.1202 | 0.5267 | | 227/358/157 | -0.1113 | 0.8119 | |
| 120 | SNP 4 | p83884-rs701106 | Intron 12 | T, 0.2597 | TT/CT/CC | 49/289/405 | 0.2471 | 0.2601 | | 51/291/412 | 1.2967 | 0.0156 | |
| 121 | SNP 1 | p82434-rs838889 | Intron 12 | C, 0.0315 | CC/TC/TT | 2/42/695 | -1.0389 | 0.0526 | 0.2565 | 2/41/702 | -0.9096 | 0.4967 | 0.1513 |
| 121 | SNP 2 | p83547-rs838887 | Intron 12 | G, 0.4564 | CC/CG/GG | 225/357/154 | 0.1202 | 0.5267 | | 227/358/157 | -0.1113 | 0.8119 | |
| 121 | SNP 3 | p83884-rs701106 | Intron 12 | T, 0.2597 | TT/CT/CC | 49/289/405 | 0.2471 | 0.2601 | | 51/291/412 | 1.2967 | 0.0156 | |
| 121 | SNP 4 | p86245-rs188375019 | Intron 12 | T, 0.0341 | CC/CT | 690/50 | 0.7447 | 0.1639 | | 696/50 | 1.8399 | 0.1674 | |
| 122 | SNP 1 | p83547-rs838887 | Intron 12 | G, 0.4564 | CC/CG/GG | 225/357/154 | 0.1202 | 0.5267 | 0.3275 | 227/358/157 | -0.1113 | 0.8119 | 0.2640 |
| 122 | SNP 2 | p83884-rs701106 | Intron 12 | T, 0.2597 | TT/CT/CC | 49/289/405 | 0.2471 | 0.2601 | | 51/291/412 | 1.2967 | 0.0156 | |
| 122 | SNP 3 | p86245-rs188375019 | Intron 12 | T, 0.0341 | CC/CT | 690/50 | 0.7447 | 0.1639 | | 696/50 | 1.8399 | 0.1674 | |
| 122 | SNP 4 | p86276-rs747155 | Intron 12 | T, 0.1495 | TT/CT/CC | 17/187/533 | 0.2793 | 0.2980 | | 17/191/541 | -0.2164 | 0.7433 | |
| 123 | SNP 1 | p83884-rs701106 | Intron 12 | T, 0.2597 | TT/CT/CC | 49/289/405 | 0.2471 | 0.2601 | **0.0386** | 51/291/412 | 1.2967 | 0.0156 | **0.0468** |
| 123 | SNP 2 | p86245-rs188375019 | Intron 12 | T, 0.0341 | CC/CT | 690/50 | 0.7447 | 0.1639 | | 696/50 | 1.8399 | 0.1674 | |
| 123 | SNP 3 | p86276-rs747155 | Intron 12 | T, 0.1495 | TT/CT/CC | 17/187/533 | 0.2793 | 0.2980 | | 17/191/541 | -0.2164 | 0.7433 | |
| 123 | SNP 4 | p86316-rs701104 | Intron 12 | T, 0.0487 | TT/GT/GG | 2/66/643 | -0.9838 | 0.0286 | | 2/64/658 | -0.6627 | 0.5579 | |
| 124 | SNP 1 | p86245-rs188375019 | Intron 12 | T, 0.0341 | CC/CT | 690/50 | 0.7447 | 0.1639 | **0.0368** | 696/50 | 1.8399 | 0.1674 | 0.2216 |
| 124 | SNP 2 | p86276-rs747155 | Intron 12 | T, 0.1495 | TT/CT/CC | 17/187/533 | 0.2793 | 0.2980 | | 17/191/541 | -0.2164 | 0.7433 | |
| 124 | SNP 3 | p86316-rs701104 | Intron 12 | T, 0.0487 | TT/GT/GG | 2/66/643 | -0.9838 | 0.0286 | | 2/64/658 | -0.6627 | 0.5579 | |
| 124 | SNP 4 | p86481-rs701103 | Exon 13-3' UTR | A, 0.2451 | AA/GA/GG | 50/259/424 | 0.1642 | 0.4492 | | 50/265/428 | -0.0074 | 0.9891 | |
| 125 | SNP 1 | p86276-rs747155 | Intron 12 | T, 0.1495 | TT/CT/CC | 17/187/533 | 0.2793 | 0.2980 | **0.0307** | 17/191/541 | -0.2164 | 0.7433 | 0.2769 |
| 125 | SNP 2 | p86316-rs701104 | Intron 12 | T, 0.0487 | TT/GT/GG | 2/66/643 | -0.9838 | 0.0286 | | 2/64/658 | -0.6627 | 0.5579 | |
| 125 | SNP 3 | p86481-rs701103 | Exon 13-3' UTR | A, 0.2451 | AA/GA/GG | 50/259/424 | 0.1642 | 0.4492 | | 50/265/428 | -0.0074 | 0.9891 | |
| 125 | SNP 4 | p86967-rs187492239 | Exon 13-3' UTR | G, 0.0355 | AA/AG | 686/52 | 0.7743 | 0.1412 | | 692/52 | 1.3818 | 0.2924 | |
| 126 | SNP 1 | p86316-rs701104 | Intron 12 | T, 0.0487 | TT/GT/GG | 2/66/643 | -0.9838 | 0.0286 | 0.1487 | 2/64/658 | -0.6627 | 0.5579 | 0.1998 |
| 126 | SNP 2 | p86481-rs701103 | Exon 13-3' UTR | A, 0.2451 | AA/GA/GG | 50/259/424 | 0.1642 | 0.4492 | | 50/265/428 | -0.0074 | 0.9891 | |
| 126 | SNP 3 | p86967-rs187492239 | Exon 13-3' UTR | G, 0.0355 | AA/AG | 686/52 | 0.7743 | 0.1412 | | 692/52 | 1.3818 | 0.2924 | |
| 126 | SNP 4 | p87011-rs58032386 | Exon 13-3' UTR | T, 0.1417 | CC/CT/TT | 544/183/14 | 0.0575 | 0.8333 | | 551/186/14 | -0.2769 | 0.6829 | |
| 127 | SNP 1 | p86481-rs701103 | Exon 13-3' UTR | A, 0.2451 | AA/GA/GG | 50/259/424 | 0.1642 | 0.4492 | 0.4750 | 50/265/428 | -0.0074 | 0.9891 | 0.8693 |
| 127 | SNP 2 | p86967-rs187492239 | Exon 13-3' UTR | G, 0.0355 | AA/AG | 686/52 | 0.7743 | 0.1412 | | 692/52 | 1.3818 | 0.2924 | |
| 127 | SNP 3 | p87011-rs58032386 | Exon 13-3' UTR | T, 0.1417 | CC/CT/TT | 544/183/14 | 0.0575 | 0.8333 | | 551/186/14 | -0.2769 | 0.6829 | |
| 127 | SNP 4 | p87266-rs150512235 | Exon 13-3' UTR[d] | C, 0.0057 | TC/TT | 9/746 | -0.0325 | 0.9789 | | 9/756 | 0.0318 | 0.9917 | |
| 128 | SNP 1 | p86967-rs187492239 | Exon 13-3' UTR | G, 0.0355 | AA/AG | 686/52 | 0.7743 | 0.1412 | 0.4515 | 692/52 | 1.3818 | 0.2924 | 0.5899 |
| 128 | SNP 2 | p87011-rs58032386 | Exon 13-3' UTR | T, 0.1417 | CC/CT/TT | 544/183/14 | 0.0575 | 0.8333 | | 551/186/14 | -0.2769 | 0.6829 | |
| 128 | SNP 3 | p87266-rs150512235 | Exon 13-3' UTR[d] | C, 0.0057 | TC/TT | 9/746 | -0.0325 | 0.9789 | | 9/756 | 0.0318 | 0.9917 | |
| 128 | SNP 4 | p87611-rs190688220 | 3' flanking | T, 0.0316 | CC/CT | 691/46 | 0.8329 | 0.1355 | | 697/46 | 1.7851 | 0.1993 | |
| 129 | SNP 1 | p87011-rs58032386 | Exon 13-3' UTR | T, 0.1417 | CC/CT/TT | 544/183/14 | 0.0575 | 0.8333 | 0.1918 | 551/186/14 | -0.2769 | 0.6829 | 0.7888 |
| 129 | SNP 2 | p87266-rs150512235 | Exon 13-3' UTR[d] | C, 0.0057 | TC/TT | 9/746 | -0.0325 | 0.9789 | | 9/756 | 0.0318 | 0.9917 | |
| 129 | SNP 3 | p87611-rs190688220 | 3' flanking | T, 0.0316 | CC/CT | 691/46 | 0.8329 | 0.1355 | | 697/46 | 1.7851 | 0.1993 | |
| 129 | SNP 4 | p87681-rs838883 | 3' flanking | A, 0.0459 | AA/GA/GG | 1/65/646 | -0.9433 | 0.0427 | | 1/62/659 | -0.0535 | 0.9639 | |
| 130 | SNP 1 | p87266-rs150512235 | Exon 13-3' UTR[d] | C, 0.0057 | TC/TT | 9/746 | -0.0325 | 0.9789 | 0.0787 | 9/756 | 0.0318 | 0.9917 | 0.5394 |
| 130 | SNP 2 | p87611-rs190688220 | 3' flanking | T, 0.0316 | CC/CT | 691/46 | 0.8329 | 0.1355 | | 697/46 | 1.7851 | 0.1993 | |
| 130 | SNP 3 | p87681-rs838883 | 3' flanking | A, 0.0459 | AA/GA/GG | 1/65/646 | -0.9433 | 0.0427 | | 1/62/659 | -0.0535 | 0.9639 | |
| 130 | SNP 4 | p87694-chr12_125261826 | 3' flanking | T, 0.0020 | CC/CT | 722/3 | 3.4021 | 0.1098 | | 731/3 | 3.0751 | 0.5620 | |
| 131 | SNP 1 | p87611-rs190688220 | 3' flanking | T, 0.0316 | CC/CT | 691/46 | 0.8329 | 0.1355 | 0.0653 | 697/46 | 1.7851 | 0.1993 | 0.6272 |
| 131 | SNP 2 | p87681-rs838883 | 3' flanking | A, 0.0459 | AA/GA/GG | 1/65/646 | -0.9433 | 0.0427 | | 1/62/659 | -0.0535 | 0.9639 | |
| 131 | SNP 3 | p87694-chr12_125261826 | 3' flanking | T, 0.0020 | CC/CT | 722/3 | 3.4021 | 0.1098 | | 731/3 | 3.0751 | 0.5620 | |
| 131 | SNP 4 | p87723-rs838881 | 3' flanking | T, 0.3183 | TT/CT/CC | 63/341/332 | 0.1390 | 0.5146 | | 65/344/338 | -0.1897 | 0.7187 | |
| 132 | SNP 1 | p87681-rs838883 | 3' flanking | A, 0.0459 | AA/GA/GG | 1/65/646 | -0.9433 | 0.0427 | 0.1085 | 1/62/659 | -0.0535 | 0.9639 | 0.9672 |
| 132 | SNP 2 | p87694-chr12_125261826 | 3' flanking | T, 0.0020 | CC/CT | 722/3 | 3.4021 | 0.1098 | | 731/3 | 3.0751 | 0.5620 | |

| | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 132 | SNP 3 | p87723-rs838881 | 3' flanking | T, 0.3183 | TT/CT/CC | 63/341/332 | 0.1390 | 0.5146 | | 65/344/338 | -0.1897 | 0.7187 | |
| 132 | SNP 4 | p87749-rs76465225 | 3' flanking | A, 0.0844 | AA/GA/GG | 7/109/620 | -0.1992 | 0.5583 | | 7/111/629 | -0.1205 | 0.8865 | |
| 133 | SNP 1 | p87694-chr12_125261826 | 3' flanking | T, 0.0020 | CC/CT | 722/3 | 3.4021 | 0.1098 | 0.5109 | 731/3 | 3.0751 | 0.5620 | 0.9769 |
| 133 | SNP 2 | p87723-rs838881 | 3' flanking | T, 0.3183 | TT/CT/CC | 63/341/332 | 0.1390 | 0.5146 | | 65/344/338 | -0.1897 | 0.7187 | |
| 133 | SNP 3 | p87749-rs76465225 | 3' flanking | A, 0.0844 | AA/GA/GG | 7/109/620 | -0.1992 | 0.5583 | | 7/111/629 | -0.1205 | 0.8865 | |
| 133 | SNP 4 | p87927-rs838880 | 3' flanking | A, 0.2414 | AA/GA/GG | 39/275/418 | 0.0198 | 0.9314 | | 40/280/423 | -0.2130 | 0.7056 | |

ApoA-I, apolipoprotein A-I; del/D, deletion; HDL-C, high-density lipoprotein cholesterol; ins/I, insertion; MA, minor allele; MAF, minor allele frequency; NA, not analyzed; SNP, single nucleotide polymorphism; UTR, untranslated region; W, wild type allele on RefSeq for insertion and deletion variations.

SNP 1-SNP 4 in each window are shown as “SNP name-SNP ID/Chr12 Position (for novel variants)” and corresponding to 5’ to 3’ direction.

All alleles on reverse strand. Splice site is defined as ± 20 bp from the start or end of an exon.

HDL-C and ApoA-I values were Box-Cox transformed.

Results were adjusted for covariates: sex, age, waist, current smoking (yes/no), and, minutes of daily walking or biking to work (jobmin) for HDL-C; sex and age for ApoA-I.

Significant global *P*-values (*P* <0.05) are shown in **bold**, see haplotype association plots in Figure 7.3.

[a, c] Based on the RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics).

[b] dbSNP build 139: GRCh37.p10; 10 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_AB).

[d] Close to the miRNA-145 seed binding site based on TargetScanHuman (version 6.2, http://www.targetscan.org/).

**Table B12. Summary of RegulomeDB scores of 153 *SCARB1* identified variants (83 sequence variants, 68 common HapMap-YRI tagSNPs, and 2 additional relevant variants from the literature).**

| RegulomeDB Score[a] | No of Variants (n) | | | | | | Total N (%) |
|---|---|---|---|---|---|---|---|
| | MAF ≥5% (n = 101) | | MAF between 1-5% (n = 27) | | MAF ≤1% (n = 25) | | |
| | Coding | Non-coding | Coding | Non-coding | Coding | Non-coding | |
| 1f | | 1 | | | | | 1 (0.65) |
| 2a | | 1 | | 1 | | 1 | 3 (1.96) |
| 2b | 1 | 4 | 1 | 1 | | | 7 (4.58) |
| 3a | 1 | 1 | | | | | 2 (1.31) |
| 3b | | 1 | | | | | 1 (0.65) |
| 4 | 1 | 18 | 1 | 10 | | 6 | 36 (23.53) |
| 5 | 1 | 40 | | 11 | 2 | 13 | 67 (43.79) |
| 6 | | 12 | | 1 | | 2 | 15 (9.80) |
| 7 | | 19 | | 1 | | 1 | 21 (13.73) |
| **Total N** | **4** | **97** | **2** | **25** | **2** | **23** | **153 (100.00)** |

MAF, minor allele frequency; SNP, single nucleotide polymorphism; YRI, Yoruba people of Ibadan, Nigeria.

A list of 83 sequence variants is shown in Table B3.

A list of 68 common HapMap-YRI tagSNPs is shown in Table B5.

[a] The RegulomeDB (version 1.0) scoring scheme represented as following: **score 1a**, expression quantitative trait loci (eQTL) + transcription factor (TF) binding + matched TF motif + matched DNase Footprint + DNase peak; **score 1b**, eQTL + TF binding + any motif + DNase Footprint + DNase peak; **score 1c**, eQTL + TF binding + matched TF motif + DNase peak; score 1d, eQTL + TF binding + any motif + DNase peak; **score 1e**, eQTL + TF binding + matched TF motif; **score 1f**, eQTL + TF binding / DNase peak, **score 2a**, TF binding + matched TF motif + matched DNase Footprint + DNase peak; **score 2b**, TF binding + any motif + DNase Footprint + DNase peak; **score 2c**, TF binding + matched TF motif + DNase peak; **score 3a**, TF binding + any motif + DNase peak; **score 3b**, TF binding + matched TF motif; **score 4**,TF binding + DNase peak; **score 5**, TF binding or DNase peak; **score 6**, others; **score 7**, no data, or can be seen at http://regulome.stanford.edu/help).

**Table B13. RegulomeDB scores and functional assignments of 153 *SCARB1* identified variants (83 sequence variants, 68 common HapMap-YRI tagSNPs, and 2 additional relevant variants from the literature).**

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | MAF | Associated Trait(s)[d] | RegulomeDB Score[e] | RegulomeDB Functional Assignment |
|---|---|---|---|---|---|---|---|---|---|---|
| p972 | rs181338950 | 125348548 | 5' flanking-promoter | | | C/T | 0.048 | | 2a | Motifs\|Footprinting\|SP1,<br>Motifs\|PWM\|Zfp410,<br>Motifs\|PWM\|SP1,<br>Motifs\|PWM\|KROX,<br>Motifs\|Footprinting\|Sp1,<br>Motifs\|Footprinting\|SP4,<br>Motifs\|PWM\|UF1H3BETA,<br>Motifs\|Footprinting\|KROX,<br>Motifs\|Footprinting\|Klf4,<br>Motifs\|PWM\|Bcl6b,<br>Motifs\|Footprinting\|UF1H3BETA,<br>Motifs\|PWM\|Klf4,<br>Motifs\|PWM\|Sp1,<br>Motifs\|PWM\|SP4,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|GATA1,<br>Protein_Binding\|ChIP-seq\|GABPA,<br>Protein_Binding\|ChIP-seq\|EGR1,<br>Protein_Binding\|ChIP-seq\|TFAP2C,<br>Protein_Binding\|ChIP-seq\|TFAP2A,<br>Protein_Binding\|ChIP-seq\|MYC,<br>Protein_Binding\|ChIP-seq\|MAX,<br>Protein_Binding\|ChIP-seq\|IRF1,<br>Protein_Binding\|ChIP-seq\|ELF1,<br>Protein_Binding\|ChIP-seq\|SP1,<br>Protein_Binding\|ChIP-seq\|CCNT2,<br>Protein_Binding\|ChIP-seq\|TBP,<br>Protein_Binding\|ChIP-seq\|YY1,<br>Protein_Binding\|ChIP-seq\|POLR2A,<br>Protein_Binding\|ChIP-seq\|TAF1,<br>Protein_Binding\|ChIP-seq\|ZBTB7A,<br>Protein_Binding\|ChIP-seq\|CTCF |
| p1048 insC (1048_ 1049) | | 125348472 | Exon 1- 5' UTR | | insC | [-/C] | 0.0079 | | 2a | Motifs\|PWM\|HIC1,<br>Motifs\|Footprinting\|AP-2gamma,<br>Motifs\|Footprinting\|TFAP2A,<br>Motifs\|Footprinting\|AP-2alpha,<br>Motifs\|Footprinting\|HIC1,<br>Motifs\|PWM\|AP-2alpha,<br>Motifs\|PWM\|AP-2gamma,<br>Motifs\|PWM\|AP-2,<br>Motifs\|PWM\|TFAP2A,<br>Motifs\|Footprinting\|AP-2,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|GATA1,<br>Protein_Binding\|ChIP-seq\|SIN3A,<br>Protein_Binding\|ChIP-seq\|HEY1,<br>Protein_Binding\|ChIP-seq\|EGR1,<br>Protein_Binding\|ChIP-seq\|TFAP2C,<br>Protein_Binding\|ChIP-seq\|TFAP2A,<br>Protein_Binding\|ChIP-seq\|IRF1,<br>Protein_Binding\|ChIP-seq\|RAD21,<br>Protein_Binding\|ChIP-seq\|SMC3,<br>Protein_Binding\|ChIP-seq\|SP1,<br>Protein_Binding\|ChIP-seq\|CCNT2,<br>Protein_Binding\|ChIP-seq\|TBP,<br>Protein_Binding\|ChIP-seq\|POLR2A,<br>Protein_Binding\|ChIP-seq\|TAF1,<br>Protein_Binding\|ChIP-seq\|CTCFL,<br>Protein_Binding\|ChIP-seq\|CTCF,<br>Protein_Binding\|ChIP-seq\|EBF1,<br>Protein_Binding\|ChIP-seq\|CDX2 |
| p1257 | rs4238001 | 125348263 | Exon 1 | Gly2Ser | | G/A | 0.032 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|GATA1,<br>Protein_Binding\|ChIP-seq\|MXI1, |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | MAF | Associated Trait(s)[d] | RegulomeDB Score[e] | RegulomeDB Functional Assignment |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | Protein_Binding\|ChIP-seq\|TFAP2C, Protein_Binding\|ChIP-seq\|TFAP2A, Protein_Binding\|ChIP-seq\|POU2F2, Protein_Binding\|ChIP-seq\|RAD21, Protein_Binding\|ChIP-seq\|CCNT2, Protein_Binding\|ChIP-seq\|E2F6, Protein_Binding\|ChIP-seq\|POLR2A, Protein_Binding\|ChIP-seq\|TAF1, Protein_Binding\|ChIP-seq\|CDX2 |
| p1265 | rs2070242 | 125348255 | Exon 1 | Ser4Ser | | C/T | 0.1284 | | 2b | Motifs\|PWM\|E2F-1, Motifs\|Footprinting\|E2F-1, Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|GATA1, Protein_Binding\|ChIP-seq\|MXI1, Protein_Binding\|ChIP-seq\|TFAP2A, Protein_Binding\|ChIP-seq\|POU2F2, Protein_Binding\|ChIP-seq\|RAD21, Protein_Binding\|ChIP-seq\|E2F6, Protein_Binding\|ChIP-seq\|POLR2A, Protein_Binding\|ChIP-seq\|TAF1, Protein_Binding\|ChIP-seq\|CDX2 |
| p1316 | rs10396208 | 125348204 | Exon 1 | Cys21Cys | | C/T | 0.0476 | | 2b | Motifs\|Footprinting\|NRSF, Motifs\|PWM\|REST, Motifs\|PWM\|NRSF, Motifs\|PWM\|Osr2, Motifs\|Footprinting\|REST, Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|GATA1, Protein_Binding\|ChIP-seq\|MXI1, Protein_Binding\|ChIP-seq\|E2F6, Protein_Binding\|ChIP-seq\|CDX2 |
| p1419 | rs201717369 | 125348101 | Intron 1 | | | G/A | 0.0121 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|GATA1, Protein_Binding\|ChIP-seq\|E2F6, Protein_Binding\|ChIP-seq\|ZBTB7A, Protein_Binding\|ChIP-seq\|E2F1 |
| p4072 | rs7139401 | 125345448 | Intron 1 | | | T/C | 0.4386 | | 3b | Motifs\|PWM\|Mafk, Motifs\|PWM\|Dlx3, Motifs\|PWM\|Dlx4, Protein_Binding\|ChIP-seq\|MAFK |
| p5055 | rs11057869 | 125344465 | Intron 1 | | | G/A | 0.0740 | | 7 | No data |
| p6600 | rs12831105 | 125342920 | Intron 1 | | | C/T | 0.1188 | | 7 | No data |
| p7650 | rs11615630 | 125341870 | Intron 1 | | | G/A | 0.0436 | | 5 | Chromatin_Structure\|DNase-seq |
| p10292 | rs4765181 | 125339228 | Intron 1 | | | G/T | 0.2490 | | 5 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq |
| p10991 | rs10773112 | 125338529 | Intron 1 | | | G/A | 0.3534 | | 4 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|FOXA1, Protein_Binding\|ChIP-seq\|RXRA, Protein_Binding\|ChIP-seq\|HDAC2 |
| p13570 | rs11057864 | 125335950 | Intron 1 | | | G/T | 0.1180 | | 4 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|HNF4A |
| p16565 | rs10773111 | 125332955 | Intron 1 | | | G/A | 0.1928 | | 6 | Single_Nucleotides\|SCARB1\|eQTL |
| p20207 | rs11057853 | 125329313 | Intron 1 | | | G/A | 0.4484 | HDL-C | 5 | Motifs\|PWM\|HNF4, Motifs\|PWM\|HNF4A, Motifs\|PWM\|HNF4directrepeat1, Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq |
| p20694 | rs11057852 | 125328826 | Intron 1 | | | G/A | 0.1050 | | 7 | No data |
| p20741 | rs11057851 | 125328779 | Intron 1 | | | C/T | 0.3237 | HDL-C (best SNP), ApoA-I | 5 | Chromatin_Structure\|DNase-seq |
| p21145 | rs3924313 | 125328375 | Intron 1 | | | C/T | 0.1772 | | 6 | Motifs\|PWM\|TFIIA |
| p22116 | rs12370382 | 125327404 | Intron 1 | | | G/A | 0.0645 | | 1f | Single_Nucleotides\|SCARB1\|eQTL, Chromatin_Structure\|DNase-seq |
| p22168 | rs7137797 | 125327352 | Intron 1 | | | T/C | 0.3977 | | 2b | Motifs\|PWM\|Staf, Motifs\|Footprinting\|Staf, Motifs\|Footprinting\|znf143, Motifs\|PWM\|znf143, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|GATA1 |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | MAF | Associated Trait(s)[d] | RegulomeDB Score[e] | RegulomeDB Functional Assignment |
|---|---|---|---|---|---|---|---|---|---|---|
| p22331 | rs6488944 | 125327189 | Intron 1 | | | T/G | 0.1474 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|GATA1,<br>Protein_Binding\|ChIP-seq\|EP300,<br>Protein_Binding\|ChIP-seq\|GATA2 |
| p22675 | rs12425134 | 125326845 | Intron 1 | | | G/T | 0.0526 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|GATA1 |
| p28137 | rs12229555 | 125321383 | Intron 1 | | | A/G | 0.3896 | | 7 | No data |
| p28692 | rs4765622 | 125320828 | Intron 1 | | | C/T | 0.2565 | | 5 | Chromatin_Structure\|DNase-seq |
| p28957 | rs11057844 | 125320563 | Intron 1 | | | G/A | 0.2362 | | 5 | Motifs\|PWM\|HTF,<br>Motifs\|Footprinting\|HTF,<br>Protein_Binding\|ChIP-seq\|GATA1 |
| p29749 | rs10846751 | 125319771 | Intron 1 | | | C/T | 0.4492 | | 7 | No data |
| p31072 | rs10846749 | 125318448 | Intron 1 | | | C/G | 0.4461 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|EP300,<br>Protein_Binding\|ChIP-seq\|MAX,<br>Protein_Binding\|ChIP-seq\|NANOG,<br>Protein_Binding\|ChIP-seq\|USF1,<br>Protein_Binding\|ChIP-seq\|TCF4,<br>Protein_Binding\|ChIP-seq\|SP1,<br>Protein_Binding\|ChIP-seq\|JUND,<br>Protein_Binding\|ChIP-seq\|TCF12,<br>Protein_Binding\|ChIP-seq\|RXRA |
| p31938 | rs10744182 | 125317582 | Intron 1 | | | G/A | 0.1837 | | 5 | Chromatin_Structure\|DNase-seq |
| p32129 | rs10773107 | 125317391 | Intron 1 | | | G/T | 0.1009 | | 7 | No data |
| p32273 | rs12580803 | 125317247 | Intron 1 | | | T/C | 0.1006 | | 5 | Chromatin_Structure\|DNase-seq |
| p32290 | rs10744181 | 125317230 | Intron 1 | | | T/C | 0.1238 | | 5 | Motifs\|Footprinting\|AR,<br>Motifs\|PWM\|AR,<br>Chromatin_Structure\|DNase-seq |
| p32395 | rs12581963 | 125317125 | Intron 1 | | | C/T | 0.1314 | | 5 | Motifs\|PWM\|E2F-1,<br>Chromatin_Structure\|DNase-seq |
| p32750 | rs7967521 | 125316770 | Intron 1 | | | A/G | 0.3425 | | 7 | No data |
| p32777 | rs11057841 | 125316743 | Intron 1 | | | G/A | 0.2805 | | 7 | No data |
| p32860 | rs7967406 | 125316660 | Intron 1 | | | A/C | 0.0991 | | 6 | Motifs\|PWM\|Hbp1,<br>Motifs\|PWM\|Rfx3 |
| p33531 | rs11057838 | 125315989 | Intron 1 | | | C/A | 0.2278 | | 7 | No data |
| p36094 | rs11608336 | 125313426 | Intron 1 | | | G/A | 0.1543 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|HNF4A,<br>Protein_Binding\|ChIP-seq\|TFAP2C,<br>Protein_Binding\|ChIP-seq\|TFAP2A,<br>Protein_Binding\|ChIP-seq\|STAT1,<br>Protein_Binding\|ChIP-seq\|GTF2F1,<br>Protein_Binding\|ChIP-seq\|TBP,<br>Protein_Binding\|ChIP-seq\|NR3C1,<br>Protein_Binding\|ChIP-seq\|STAT3,<br>Protein_Binding\|ChIP-seq\|ESR1,<br>Protein_Binding\|ChIP-seq\|POLR2A,<br>Protein_Binding\|ChIP-seq\|CDX2 |
| p36461 | rs4765178 | 125313059 | Intron 1 | | | C/T | 0.1671 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|HNF4A,<br>Protein_Binding\|ChIP-seq\|TFAP2A,<br>Protein_Binding\|ChIP-seq\|NR3C1,<br>Protein_Binding\|ChIP-seq\|ESR1 |
| p36908 | rs10846745 | 125312612 | Intron 1 | | | C/G | 0.3257 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|HNF4A,<br>Protein_Binding\|ChIP-seq\|EP300,<br>Protein_Binding\|ChIP-seq\|TFAP2C,<br>Protein_Binding\|ChIP-seq\|AR,<br>Protein_Binding\|ChIP-seq\|FOXA1,<br>Protein_Binding\|ChIP-seq\|CDX2 |
| p37095 | rs10846744 | 125312425 | Intron 1 | | | C/G | 0.3056 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|HNF4A,<br>Protein_Binding\|ChIP-seq\|EP300,<br>Protein_Binding\|ChIP-seq\|TFAP2C,<br>Protein_Binding\|ChIP-seq\|MAX,<br>Protein_Binding\|ChIP-seq\|AR,<br>Protein_Binding\|ChIP-seq\|ESR1,<br>Protein_Binding\|ChIP-seq\|FOXA1,<br>Protein_Binding\|ChIP-seq\|CDX2, |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | MAF | Associated Trait(s)[d] | RegulomeDB Score[e] | RegulomeDB Functional Assignment |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | Protein_Binding\|ChIP-seq\|GATA3 |
| p41632 | rs6488943 | 125307888 | Intron 1 | | | A/C | 0.2954 | | 5 | Chromatin_Structure\|DNase-seq |
| p42467 | rs11057830 | 125307053 | Intron 1 | | | C/T | 0.1523 | | 7 | No data |
| p45516 | rs1902569 | 125304004 | Intron 1 | | | G/A | 0.1544 | HDL-C | 5 | Chromatin_Structure\|DNase-seq |
| p45627 | rs12297372 | 125303893 | Intron 1 | | | A/G | 0.0487 | | 5 | Chromatin_Structure\|DNase-seq |
| p46964 | rs114061302 | 125302556 | Intron 1 | | | G/A | 0.0388 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|IKZF1 |
| p48969 | rs2343394 | 125300551 | Intron 2 | | | C/T | 0.1898 | | 5 | Chromatin_Structure\|DNase-seq |
| p49537 | rs7305310 | 125299983 | Intron 2 | | | C/T | 0.1007 | | 5 | Chromatin_Structure\|DNase-seq |
| p49570 delC | rs145376237 | 125299950 | Intron 2 | | delC | [C/-] | 0.2276 | | 5 | Motifs\|PWM\|Ascl2,<br>Motifs\|PWM\|E47,<br>Motifs\|Footprinting\|E47,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p49690 | rs4765615 | 125299830 | Intron 2 | | | G/A | 0.4426 | HDL-C, ApoA-I | 5 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p49759 | rs146272788 | 125299761 | Intron 2 | | | C/T | 0.0020 | | 5 | Motifs\|PWM\|Gm397,<br>Motifs\|PWM\|Zscan4,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p49978 | rs5891 | 125299542 | Exon 3 | Val135Ile | | G/A | 0.0058 | | 5 | Motifs\|Footprinting\|Pax-3,<br>Motifs\|PWM\|Pax-3,<br>Chromatin_Structure\|DNase-seq |
| p50024 | rs368880622 | 125299496 | Intron 3 | | | G/T | 0.0026 | | 5 | Chromatin_Structure\|DNase-seq |
| p50118 | rs58710319 | 125299402 | Intron 3 | | | C/T | 0.0208 | | 5 | Chromatin_Structure\|DNase-seq |
| p50151 | rs2278986 | 125299369 | Intron 3 | | | T/C | 0.1933 | | 5 | Chromatin_Structure\|DNase-seq |
| p50380 | rs141748317 | 125299140 | Intron 3 | | | A/G | 0.0112 | | 2b | Motifs\|Footprinting\|HEN1,<br>Motifs\|PWM\|HEN1,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|IKZF1 |
| p50489 | rs61320152 | 125299031 | Intron 3 | | | G/T | 0.0257 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|IKZF1 |
| p50954 | | 125298566 | Intron 4 | | | T/C | 0.0007 | | 5 | Chromatin_Structure\|DNase-seq |
| p51888 | rs7138304 | 125297632 | Intron 4 | | | C/T | 0.1079 | | 2b | Motifs\|Footprinting\|TRF1,<br>Motifs\|PWM\|TRF1,<br>Motifs\|Footprinting\|IRF-1,<br>Motifs\|PWM\|IRF-1,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|NFYB,<br>Protein_Binding\|ChIP-seq\|USF2,<br>Protein_Binding\|ChIP-seq\|USF1,<br>Protein_Binding\|ChIP-seq\|IKZF1 |
| p52096 | rs10846739 | 125297424 | Intron 4 | | | A/G | 0.4693 | | 3a | Motifs\|PWM\|Tbp,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|IKZF1 |
| p52556 | rs11057820 | 125296964 | Intron 4 | | | G/A | 0.1000 | | 5 | Chromatin_Structure\|DNase-seq |
| p52610 | rs10846738 | 125296910 | Intron 4 | | | C/T | 0.1349 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|SPI1 |
| p52919 | | 125296601 | Intron 4 | | | G/T | 0.0013 | | 5 | Motifs\|PWM\|Zfp281,<br>Motifs\|PWM\|MZF1_5-13,<br>Motifs\|Footprinting\|MZF1_5-13,<br>Motifs\|PWM\|Gabpa,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p52956 | rs77740046 | 125296564 | Intron 4 | | | C/T | 0.0546 | | 5 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p52995 | rs113910315 | 125296525 | Intron 4-splice site | | | T/G | 0.0020 | | 5 | Motifs\|PWM\|Zic3, Motifs\|PWM\|Zic2,<br>Motifs\|PWM\|Zic1,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p53128 | rs372212527 | 125296392 | Intron 5 | | | C/T | 0.005 | | 5 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p53159 | rs59809936 | 125296361 | Intron 5 | | | G/A | 0.049 | | 5 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p53359 | rs112371713 | 125296161 | Intron 5 | | | G/A | 0.1243 | | 5 | Chromatin_Structure\|DNase-seq |
| p53372 | rs115604379 | 125296148 | Intron 5 | | | C/T | 0.0066 | | 5 | Chromatin_Structure\|DNase-seq |
| p53481 | rs143611171 | 125296039 | Intron 5 | | | C/T | 0.099 | | 6 | Motifs\|Footprinting\|ARP-1(COUP-TF2), Motifs\|PWM\|ARP-1(COUP-TF2) |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | MAF | Associated Trait(s)[d] | RegulomeDB Score[e] | RegulomeDB Functional Assignment |
|---|---|---|---|---|---|---|---|---|---|---|
| p53790 | rs4765614 | 125295730 | Intron 5 | | | G/A | 0.2653 | | 5 | Chromatin_Structure\|DNase-seq |
| p54445 | rs60910935 | 125295075 | Intron 5 | | | A/G | 0.0418 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|RFX3 |
| p54475 | rs60227139 | 125295045 | Intron 5 | | | C/T | 0.0437 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|RFX3 |
| p54492 | rs61762481 | 125295028 | Intron 5 | | | G/A | 0.1005 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|RFX3 |
| p54611 | | 125294909 | Intron 5 | | | T/C | 0.0007 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|RFX3 |
| p54627 | | 125294893 | Intron 5 | | | G/C | 0.0020 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|RFX3 |
| p54856 | | 125294664 | Intron 6 | | | C/T | 0.0007 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|RFX3, Protein_Binding\|ChIP-seq\|STAT3 |
| p55923 | rs838900 | 125293597 | Intron 6 | | | G/A | 0.3921 | | 7 | No data |
| p55963 | rs7134858 | 125293557 | Intron 6 | | | C/T | 0.1560 | ApoA-I (best SNP) | 6 | Motifs\|PWM\|STAT1 |
| p56845 | rs838902 | 125292675 | Intron 6 | | | A/G | 0.4249 | | 5 | Motifs\|PWM\|Bbx, Chromatin_Structure\|DNase-seq |
| p57004 | rs187562853 | 125292516 | Intron 6 | | | G/A | 0.0098 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|GATA1 |
| p57107 | rs5892 | 125292413 | Exon 7 | Phe301Phe | | C/T | 0.0589 | | 4 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|GATA1, Protein_Binding\|ChIP-seq\|NR3C1 |
| p57508 | rs71458866 | 125292012 | Intron 7 | | | G/A | 0.1130 | | 4 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|GATA1 |
| p57592 | rs838903 | 125291928 | Intron 7 | | | G/A | 0.3763 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|GATA1 |
| p58514 | rs838905 | 125291006 | Intron 7 | | | T/C | 0.4329 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|CEBPB |
| p58664 | rs865716 | 125290856 | Intron 7 | | | A/T | 0.2708 | | 5 | Chromatin_Structure\|DNase-seq |
| p60255 | rs3782287 | 125289265 | Intron 7 | | | C/T | 0.2831 | | 5 | Chromatin_Structure\|DNase-seq |
| p61872 | rs838909 | 125287648 | Intron 7 | | | C/T | 0.2199 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|RFX3 |
| p62140 | rs838910 | 125287380 | Intron 7 | | | G/T | 0.3047 | | 5 | Chromatin_Structure\|FAIRE, Protein_Binding\|ChIP-seq\|RFX3 |
| p62409 | rs838911 | 125287111 | Intron 7 | | | C/T | 0.4211 | | 5 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq |
| p62615 | rs7138386 | 125286905 | Intron 7 | | | T/C | 0.1137 | | 5 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq |
| p63483 | rs838912 | 125286037 | Intron 7 | | | G/A | 0.0867 | ApoA-I | 7 | No data |
| p64772 | rs5888 | 125284748 | Exon 8 | Ala350Ala | | C/T | 0.0961 | ApoA-I | 3a | Motifs\|PWM\|Nr2f2, Motifs\|PWM\|Rara, Motifs\|PWM\|PPARalpha:RXRalpha, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|RFX3, Protein_Binding\|ChIP-seq\|GATA2 |
| p64923 | rs838915 | 125284597 | Intron 8 | | | C/A | 0.1435 | | 5 | Motifs\|PWM\|AP-2, Chromatin_Structure\|DNase-seq |
| p65999 | rs12819677 | 125283521 | Intron 8 | | | G/A | 0.2813 | | 6 | Motifs\|PWM\|PPARG |
| p67439 | rs961170 | 125282081 | Intron 8 | | | G/A | 0.0893 | | 4 | Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|SPI1 |
| p67700 | rs1726374 | 125281820 | Intron 8 | | | G/A | 0.1933 | | 7 | No data |
| p69013 | rs7135117 | 125280507 | Intron 8 | | | A/G | 0.2901 | | 7 | No data |
| p69699 | rs10396210 | 125279821 | Intron 8-splice site | | | G/A | 0.1511 | | 4 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|NFKB1, Protein_Binding\|ChIP-seq\|TCF12, Protein_Binding\|ChIP-seq\|POLR2A, Protein_Binding\|ChIP-seq\|EBF1 |
| p69994 | rs188094006 | 125279526 | Intron 9 | | | C/T | 0.005 | | 5 | Motifs\|PWM\|RREB-1, Motifs\|PWM\|GR, Chromatin_Structure\|DNase-seq |
| p69995 delC | rs5801571 | 125279525 | Intron 9 | | delC | [C/-] | 0.2761 | | 5 | Motifs\|PWM\|RREB-1, Motifs\|PWM\|GR, Chromatin_Structure\|DNase-seq |
| p70088 | rs117585141 | 125279432 | Intron 9 | | | G/A | 0.158 | | 7 | No data |
| p70129 | rs1814575 | 125279391 | Intron 9 | | | C/T | 0.047 | | 7 | No data |
| p70148 | rs183061101 | 125279372 | Intron 9 | | | T/C | 0.011 | | 6 | Motifs\|PWM\|Nrf-2, Motifs\|PWM\|LUN-1 |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | MAF | Associated Trait(s)[d] | RegulomeDB Score[e] | RegulomeDB Functional Assignment |
|---|---|---|---|---|---|---|---|---|---|---|
| p70201 |  | 125279319 | Intron 9 |  |  | T/C | 0.0010 |  | 6 | Motifs\|PWM\|MEF2A |
| p70292 | rs150388176 | 125279228 | Intron 9 |  |  | C/T | 0.016 |  | 5 | Chromatin_Structure\|DNase-seq |
| p71867 | rs7954022 | 125277653 | Intron 9 |  |  | C/T | 0.1323 |  | 5 | Motifs\|PWM\|EWSR1-FLI1, Motifs\|Footprinting\|EWSR1-FLI1, Chromatin_Structure\|DNase-seq |
| p72197 | rs838861 | 125277323 | Intron 9 |  |  | A/G | 0.3777 |  | 7 | No data |
| p72777 | rs838862 | 125276743 | Intron 9 |  |  | C/T | 0.0887 |  | 5 | Chromatin_Structure\|DNase-seq |
| p75766 | rs838866 | 125273754 | Intron 9 |  |  | T/C | 0.2116 |  | 6 | Single_Nucleotides\|SCARB1\|eQTL |
| p75778 | rs7301120 | 125273742 | Intron 9 |  |  | C/T | 0.1135 |  | 6 | Single_Nucleotides\|SCARB1\|eQTL |
| p76204 | rs866793 | 125273316 | Intron 9 |  |  | A/G | 0.0520 |  | 7 | No data |
| p76757 | rs9919713 | 125272763 | Intron 9 |  |  | A/T | 0.4390 |  | 6 | Motifs\|PWM\|FOXJ2, Motifs\|PWM\|Alx-4, Motifs\|Footprinting\|FOXJ2, Motifs\|Footprinting\|Alx-4 |
| p77181 | rs146246031 | 125272339 | Intron 9 |  |  | T/C | 0.0053 |  | 7 | No data |
| p77250 | rs201901986 | 125272270 | Intron 9 |  |  | G/T | 0.095 |  | 6 | Motifs\|PWM\|Srf, Motifs\|PWM\|Mtf1, Motifs\|PWM\|Tcfap2e |
| p77251 | rs34339961 | 125272269 | Intron 9 |  |  | A/T | 0.1177 |  | 6 | Motifs\|PWM\|Srf, Motifs\|PWM\|Mtf1, Motifs\|PWM\|Tcfap2e |
| p77381 | rs138499966 | 125272139 | Intron 9 |  |  | T/C | 0.0046 |  | 6 | Motifs\|PWM\|Foxl1, Motifs\|PWM\|Freac-2, Motifs\|PWM\|Foxj3, Motifs\|PWM\|FOXO3, Motifs\|PWM\|HFH8(FOXF1A), Motifs\|PWM\|FOXJ2, Motifs\|PWM\|Freac-4, Motifs\|Footprinting\|Foxq1, Motifs\|PWM\|Foxa2, Motifs\|PWM\|HFH1(FOXQ1), Motifs\|PWM\|Foxk1, Motifs\|Footprinting\|HFH1(FOXQ1), Motifs\|PWM\|Foxq1, Motifs\|PWM\|HNF3 |
| p77620 | rs377124254 | 125271900 | Intron 10 |  |  | G/A | 0.0007 |  | 5 | Chromatin_Structure\|DNase-seq |
| p77682 | rs150082885 | 125271838 | Intron 10 |  |  | A/G | 0.0106 |  | 5 | Chromatin_Structure\|DNase-seq |
| p77704 |  | 125271816 | Intron 10 |  |  | C/A | 0.0040 |  | 5 | Chromatin_Structure\|DNase-seq |
| p77842 | rs2272310 | 125271678 | Intron 10 |  |  | G/A | 0.0807 |  | 5 | Chromatin_Structure\|DNase-seq |
| p78255 | rs184052375 | 125271265 | Intron 10 |  |  | A/G | 0.0072 |  | 4 | Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq, Protein_Binding\|ChIP-seq\|SIX5, Protein_Binding\|ChIP-seq\|MAX, Protein_Binding\|ChIP-seq\|NANOG, Protein_Binding\|ChIP-seq\|USF2, Protein_Binding\|ChIP-seq\|USF1, Protein_Binding\|ChIP-seq\|ATF3, Protein_Binding\|ChIP-seq\|YY1, Protein_Binding\|ChIP-seq\|TBP, Protein_Binding\|ChIP-seq\|POLR2A, Protein_Binding\|ChIP-seq\|TAF1 |
| p78402 | rs838898 | 125271118 | Intron 10 |  |  | G/A | 0.0714 |  | 5 | Motifs\|PWM\|Hic1, Motifs\|PWM\|Eomes, Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq |
| p78430 | rs838897 | 125271090 | Intron 10 |  |  | C/G | 0.3830 |  | 5 | Motifs\|PWM\|RNF96, Motifs\|PWM\|AP-2, Motifs\|Footprinting\|AP-2, Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq |
| p78747 | rs2293440 | 125270773 | Intron 11 |  |  | T/C | 0.4112 |  | 5 | Chromatin_Structure\|DNase-seq |
| p78791 | rs75289200 | 125270729 | Intron 11 |  |  | T/C | 0.0321 |  | 5 | Chromatin_Structure\|DNase-seq |
| p79721 | rs838896 | 125269799 | Intron 11 |  |  | G/C | 0.3104 | ApoA-I | 5 | Chromatin_Structure\|DNase-seq |
| p79828 | rs838895 | 125269692 | Intron 11 |  |  | C/G | 0.3171 | ApoA-I, HDL-C | 5 | Motifs\|PWM\|Pax-5, Chromatin_Structure\|DNase-seq |
| p80045 | rs838893 | 125269475 | Intron 11 |  |  | G/A | 0.3244 |  | 5 | Chromatin_Structure\|DNase-seq |
| p81863 | rs185445624 | 125267657 | Intron 11 |  |  | G/A | 0.0020 |  | 5 | Motifs\|PWM\|ESR1, Motifs\|PWM\|Rfx3, Chromatin_Structure\|FAIRE, Chromatin_Structure\|DNase-seq |
| p82019 | rs838890 | 125267501 | Intron 11 |  |  | C/T | 0.0320 |  | 5 | Chromatin_Structure\|FAIRE, |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | MAF | Associated Trait(s)[d] | RegulomeDB Score[e] | RegulomeDB Functional Assignment |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | Chromatin_Structure\|DNase-seq |
| p82264 | rs141545424 | 125267256 | Exon 12 | Gly501Gly | | C/A | 0.0007 | | 5 | Chromatin_Structure\|DNase-seq |
| p82340 | rs77483223 | 125267180 | Intron 12 | | | G/A | 0.0231 | | 5 | Motifs\|PWM\|Arid3a,<br>Chromatin_Structure\|DNase-seq |
| p82369 | rs75446635 | 125267151 | Intron 12 | | | G/A | 0.0059 | | 5 | Chromatin_Structure\|DNase-seq |
| p82434 | rs838889 | 125267086 | Intron 12 | | | T/C | 0.0315 | | 5 | Motifs\|PWM\|FOXP1,<br>Motifs\|Footprinting\|HFH3(FOXI1),<br>Motifs\|PWM\|MRF-2,<br>Motifs\|Footprinting\|MRF-2,<br>Motifs\|PWM\|HFH3(FOXI1),<br>Chromatin_Structure\|DNase-seq |
| p83547 | rs838887 | 125265973 | Intron 12 | | | C/G | 0.4564 | | 5 | Chromatin_Structure\|DNase-seq |
| p83884 | rs701106 | 125265636 | Intron 12 | | | C/T | 0.2597 | ApoA-I | 5 | Chromatin_Structure\|DNase-seq |
| p86245 | rs188375019 | 125263275 | Intron 12 | | | C/T | 0.0341 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|BATF |
| p86276 | rs747155 | 125263244 | Intron 12 | | | C/T | 0.1495 | | 2b | Motifs\|PWM\|NGFI-C,<br>Motifs\|Footprinting\|NGFI-C,<br>Motifs\|Footprinting\|Sp3,<br>Motifs\|PWM\|Sp3,<br>Motifs\|PWM\|KROX,<br>Motifs\|PWM\|UF1H3BETA,<br>Motifs\|Footprinting\|KROX,<br>Motifs\|Footprinting\|UF1H3BETA,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|BATF |
| p86316 | rs701104 | 125263204 | Intron 12 | | | G/T | 0.0487 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|EGR1,<br>Protein_Binding\|ChIP-seq\|BATF |
| p86481 | rs701103 | 125263039 | Exon 13-3' UTR | Gly499Arg (isoform 2) | | G/A | 0.2451 | | 5 | Chromatin_Structure\|DNase-seq |
| p86967 | rs187492239 | 125262553 | Exon 13-3' UTR | | | A/G | 0.0355 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|HNF4A,<br>Protein_Binding\|ChIP-seq\|HEY1,<br>Protein_Binding\|ChIP-seq\|TCF4,<br>Protein_Binding\|ChIP-seq\|JUND,<br>Protein_Binding\|ChIP-seq\|POLR2A,<br>Protein_Binding\|ChIP-seq\|HNF4G,<br>Protein_Binding\|ChIP-seq\|FOSL2,<br>Protein_Binding\|ChIP-seq\|HDAC2 |
| p87011 | rs58032386 | 125262509 | Exon 13-3' UTR | | | C/T | 0.1417 | | 2a | Motifs\|PWM\|HNF4alpha1,<br>Motifs\|Footprinting\|HNF4alpha1,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|HNF4A,<br>Protein_Binding\|ChIP-seq\|HEY1,<br>Protein_Binding\|ChIP-seq\|TCF4,<br>Protein_Binding\|ChIP-seq\|JUND,<br>Protein_Binding\|ChIP-seq\|POLR2A,<br>Protein_Binding\|ChIP-seq\|HNF4G,<br>Protein_Binding\|ChIP-seq\|FOSL2,<br>Protein_Binding\|ChIP-seq\|HDAC2 |
| p87195 | rs10396211 | 125262325 | Exon 13-3' UTR | | | G/C | 0.122 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|TCF4 |
| p87266 | rs150512235 | 125262254 | Exon 13-3' UTR[f] | | | T/C | 0.0057 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|ELF1 |
| p87416 | rs838884 | 125262104 | 3' flanking | | | C/T | 0.301 | | 2b | Motifs\|PWM\|DEAF1,<br>Motifs\|Footprinting\|DEAF1,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|CEBPB,<br>Protein_Binding\|ChIP-seq\|ELF1,<br>Protein_Binding\|ChIP-seq\|FOXA2,<br>Protein_Binding\|ChIP-seq\|JUND,<br>Protein_Binding\|ChIP-seq\|FOSL2,<br>Protein_Binding\|ChIP-seq\|FOXA1 |
| p87459 | | 125262061 | 3' flanking | | | C/T | 0.005 | | 4 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|CEBPB,<br>Protein_Binding\|ChIP-seq\|ELF1,<br>Protein_Binding\|ChIP-seq\|FOXA2,<br>Protein_Binding\|ChIP-seq\|JUND, |

| SNP Name[a] | SNP ID[b] | Chr12 Position[c] | Location | Amino Acid Change | Indels | Major/ Minor Alleles | MAF | Associated Trait(s)[d] | RegulomeDB Score[e] | RegulomeDB Functional Assignment |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | Protein_Binding\|ChIP-seq\|FOSL2,<br>Protein_Binding\|ChIP-seq\|FOXA1 |
| p87611 | rs190688220 | 125261909 | 3' flanking | | | C/T | 0.0316 | | 4 | Chromatin_Structure\|DNase-seq,<br>Protein_Binding\|ChIP-seq\|CEBPB,<br>Protein_Binding\|ChIP-seq\|JUND,<br>Protein_Binding\|ChIP-seq\|FOSL2 |
| p87681 | rs838883 | 125261839 | 3' flanking | | | G/A | 0.0459 | | 5 | Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p87694 | | 125261826 | 3' flanking | | | C/T | 0.0020 | | 5 | Motifs\|PWM\|TP53,<br>Motifs\|PWM\|p53,<br>Chromatin_Structure\|FAIRE,<br>Chromatin_Structure\|DNase-seq |
| p87707 | rs838882 | 125261813 | 3' flanking | | | G/A | 0.303 | | 7 | No data |
| p87723 | rs838881 | 125261797 | 3' flanking | | | C/T | 0.3183 | | 6 | Motifs\|PWM\|NFIL3,<br>Motifs\|Footprinting\|NFIL3,<br>Motifs\|PWM\|TFIIA,<br>Chromatin_Structure\|FAIRE |
| p87749 | rs76465225 | 125261771 | 3' flanking | | | G/A | 0.0844 | | 7 | No data |
| p87927 | rs838880 | 125261593 | 3' flanking | | | G/A | 0.2414 | | 5 | Chromatin_Structure\|DNase-seq |

ApoA-I, apolipoprotein A-I; HDL-C, high-density lipoprotein cholesterol; del, deletion; eQTL, expression quantitative trait loci; Indels, insertion and deletion variations; ins, insertion; MAF, minor allele frequency; SNP, single nucleotide polymorphism; UTR, untranslated region; YRI, Yoruba people of Ibadan, Nigeria.

All alleles on reverse strand. Splice site is defined as ± 20 bp from the start or end of an exon.

A list of 83 sequence variants is shown in Table B3.

A list of 68 common HapMap-YRI tagSNPs is shown in Table B5.

A list of 138 genotyped variants in the entire sample of 788 African Blacks is shown in Table B6.

Underlined MAFs were derived from sequencing data due to genotyping failure (Table B3).

Double underlined MAFs were identified in sequencing stage but not selected for genotyping based on the linkage disequilibrium structure (Table B3; Figures B1 and B2). See details of selected variants in Tables B3 and B5.

[a, c] Based on the RefSeq *SCARB1*: hg19, NM_005505 (CHIP Bioinformatics).

[b] dbSNP build 139: GRCh37.p10; 10 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_AB).

[d] Evidence is based on SNPs with MAF ≥5% exhibiting nominally significant association with either HDL-C or ApoA-I (*P* <0.05; Tables B9 and B10) in single-site analysis observed in the current study.

[e] Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table B12 or can be seen at http://regulome.stanford.edu/help.

[f] Close to the miRNA-145 seed binding site based on TargetScanHuman (version 6.2, http://www.targetscan.org/).

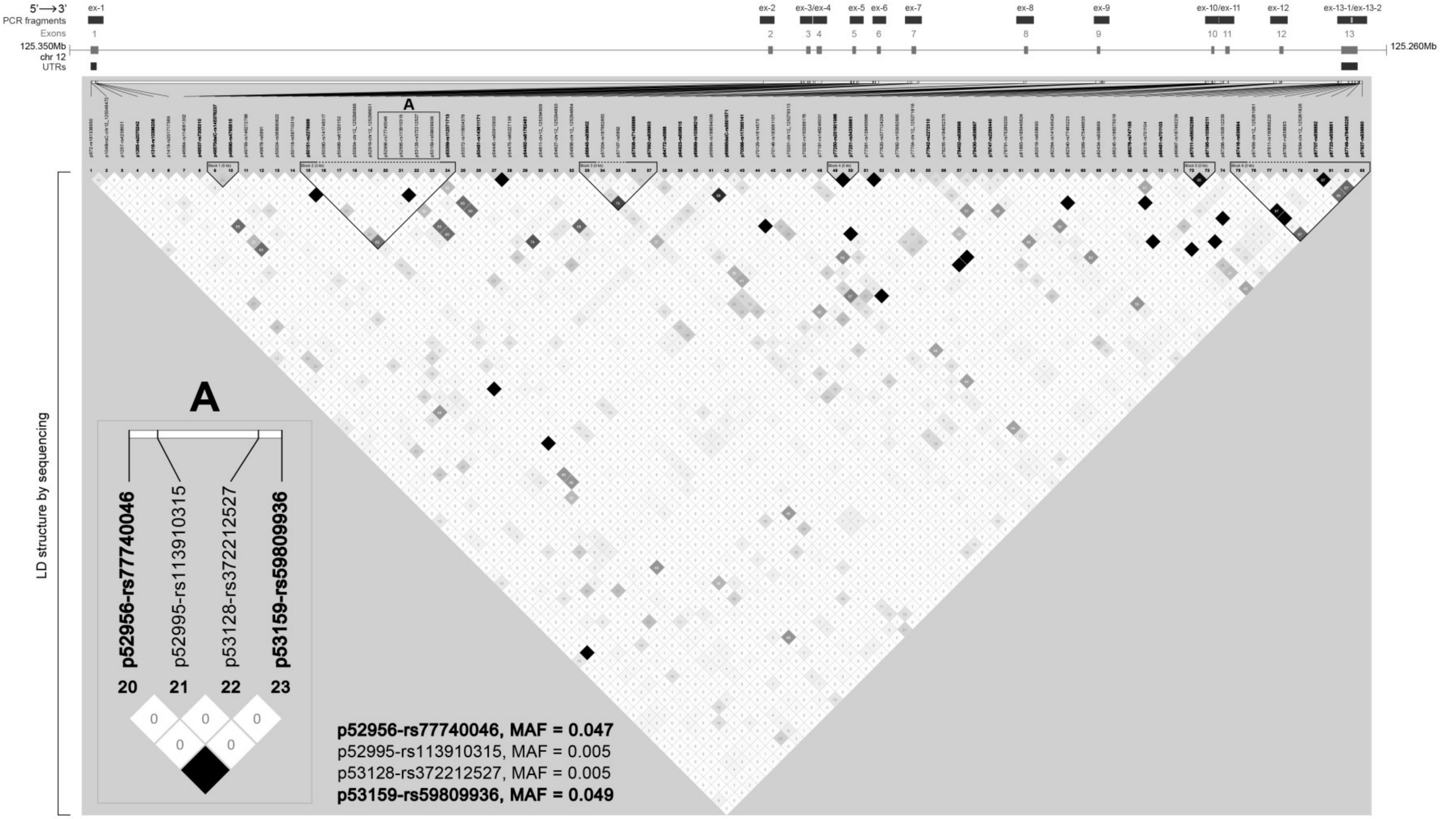

**Figure B1. Linkage disequilibrium (LD) structure of 83 *SCARB1* sequence variants.**

Of 83 *SCARB1* sequence variants (see the list in **Table B3**), 78 were selected for genotyping. An enlarged view of the part of LD plot (**A**) shows the pairwise correlations ($r^2$) between four variants including the two variants (shown in **bold**) in the same bin in our data, of which one selected for genotyping. This bin was not identified by Haploview Tagger analysis of common *SCARB1* variants in the HapMap-YRI data (see **Table B5** and **Figure B3**). MAF, minor allele frequency. YRI, Yoruba people of Ibadan, Nigeria.

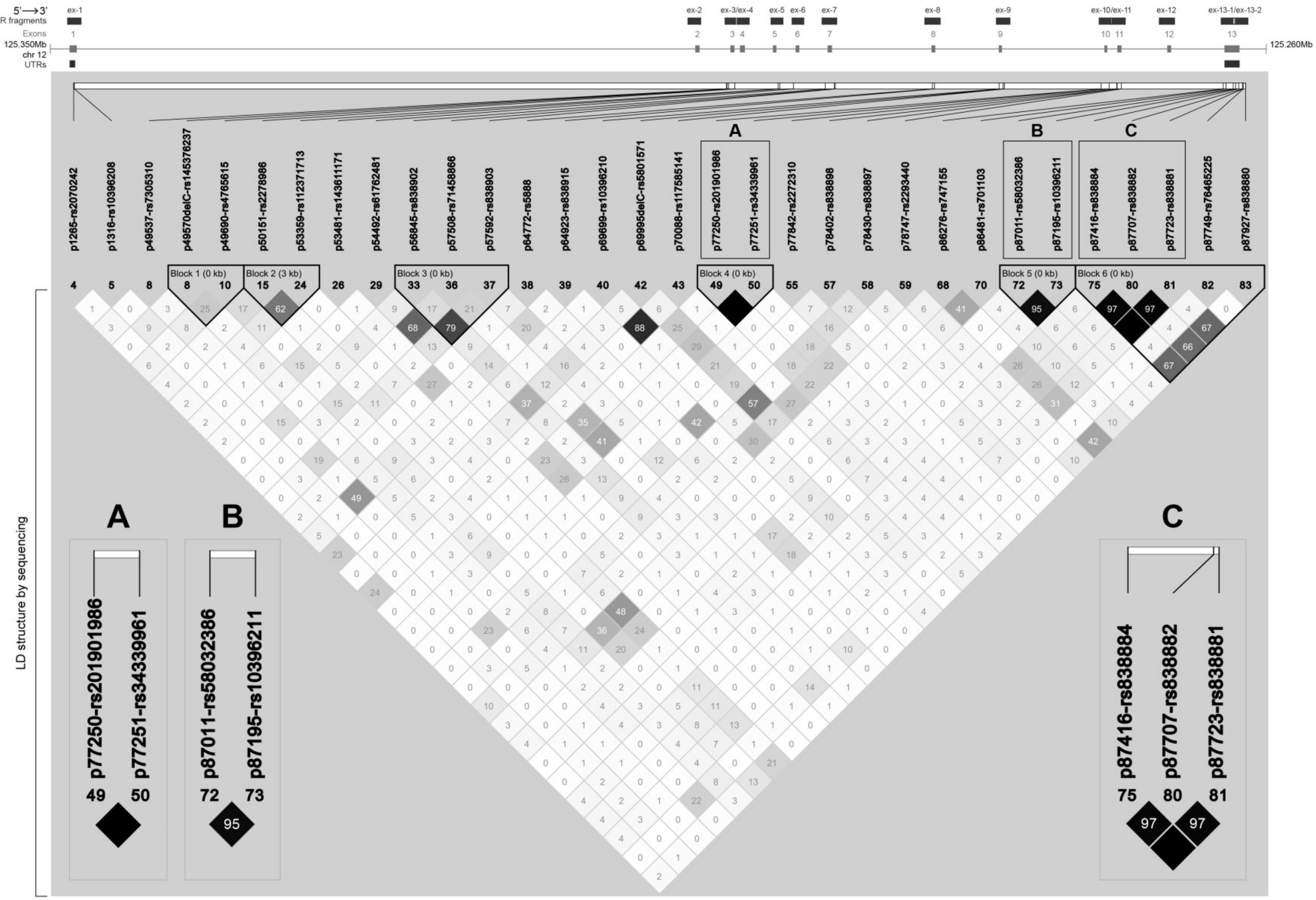
5'⟶3'
PCR fragments
Exons
125.350Mb
chr 12
UTRs
125.260Mb
ex-1
ex-2
ex-3/ex-4
ex-5
ex-6
ex-7
ex-8
ex-9
ex-10/ex-11
ex-12
ex-13-1/ex-13-2
1 2 3 4 5 6 7 8 9 10 11 12 13
A
B
C
p1265-rs2070242
p1316-rs10396208
p49537-rs7305310
p49570delC-rs145376237
p49690-rs4765615
p50151-rs2278986
p53359-rs112371713
p53481-rs143611171
p54492-rs61762481
p56845-rs838902
p57508-rs71458866
p57592-rs838903
p64772-rs5888
p64923-rs838915
p69699-rs10396210
p69995delC-rs5801571
p70088-rs117585141
p77250-rs201901986
p77251-rs34339961
p77842-rs2272310
p78402-rs838898
p78430-rs838897
p78747-rs2293440
p86276-rs747155
p86481-rs701103
p87011-rs58032386
p87195-rs10396211
p87416-rs838884
p87707-rs838882
p87723-rs838881
p87749-rs76465225
p87927-rs838880
Block 1 (0 kb)
Block 2 (3 kb)
Block 3 (0 kb)
Block 4 (0 kb)
Block 5 (0 kb)
Block 6 (0 kb)
4 5 8 8 10 15 24 26 29 33 36 37 38 39 40 42 43 49 50 55 57 58 59 68 70 72 73 75 80 81 82 83
LD structure by sequencing
A
p77250-rs201901986
p77251-rs34339961
49 50
B
p87011-rs58032386
p87195-rs10396211
72 73
95
C
p87416-rs838884
p87707-rs838882
p87723-rs838881
75 80 81
97 97

**Figure B2. Linkage disequilibrium (LD) structure of 32 *SCARB1* common sequence variants.**

Enlarged view of the parts of the LD plot (**A**, **B**, and **C**) show 3 LD bins, which were identified by Haploview Tagger analysis of variants with minor allele frequency ≥5% and $r^2$ ≥0.90, containing more than one variant ($r^2$ ranging between 0.95-1.0).

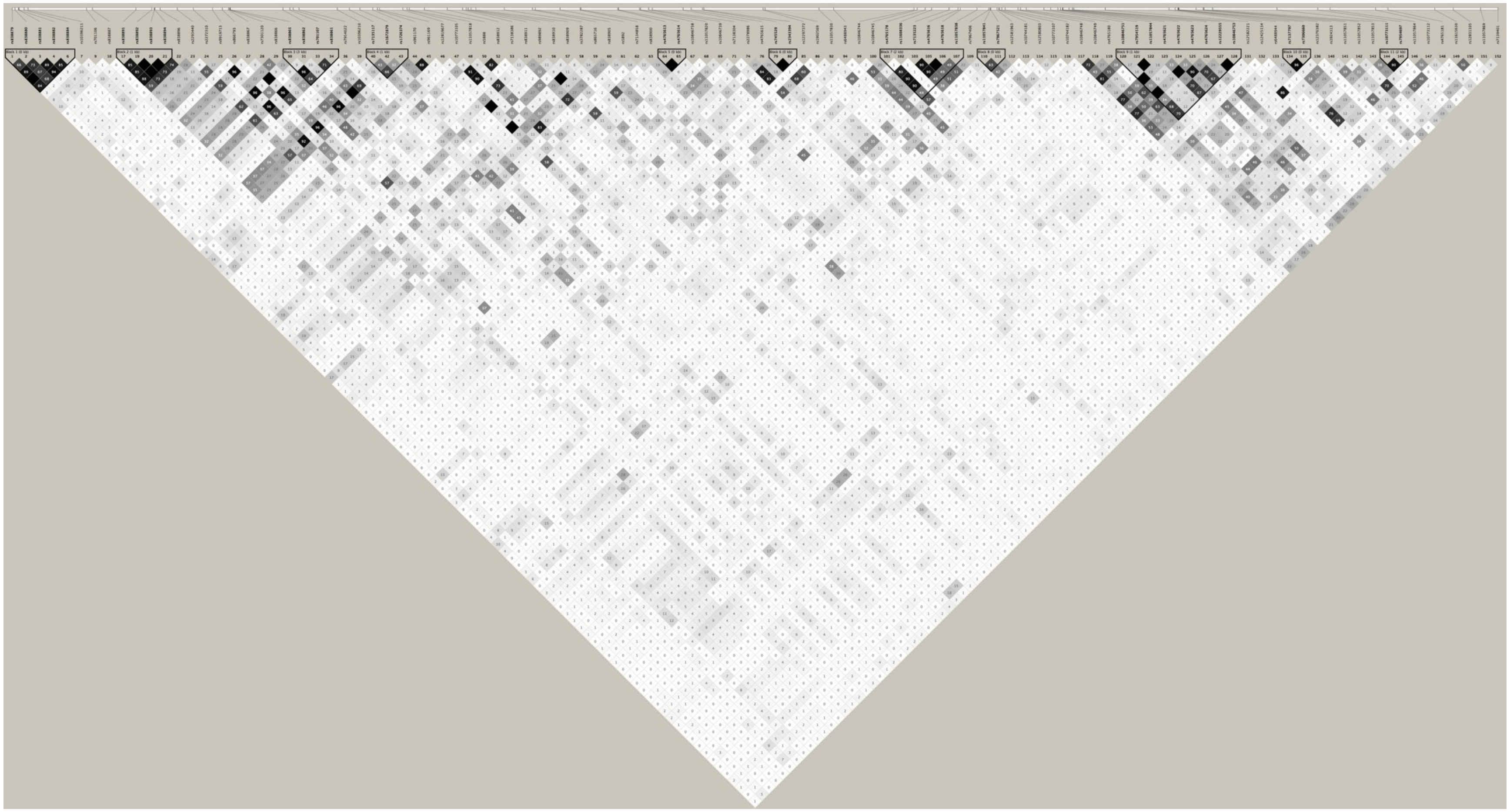

**Figure B3.** **Linkage disequilibrium structure of 108 *SCARB1* common HapMap-YRI tagSNPs.**

A list of 77 common HapMap-YRI tagSNPs identified by Haploview Tagger analysis of variants with minor allele frequency ≥5% and $r^2$ ≥0.80 is shown in **Table B5**. SNP, single nucleotide polymorphism; YRI, Yoruba people of Ibadan, Nigeria.

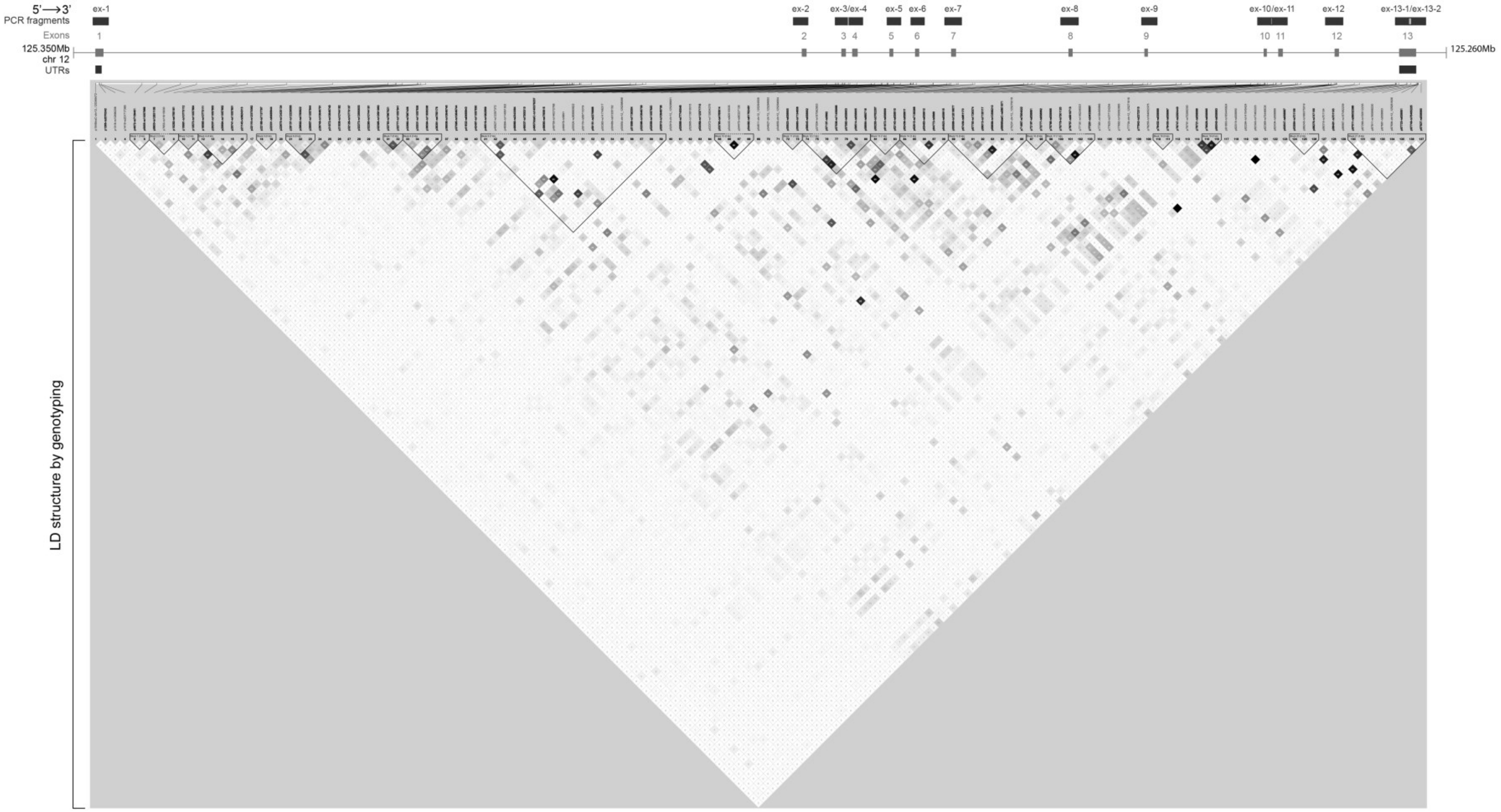

**Figure B4. Linkage disequilibrium structure of 137 *SCARB1* genotyped variants.**

A list of 87 genotyped common tagSNPs identified by Haploview Tagger analysis of variants with minor allele frequency ≥5% and $r^2$ ≥0.90 is shown in **Table B7**. SNP, single nucleotide polymorphism.

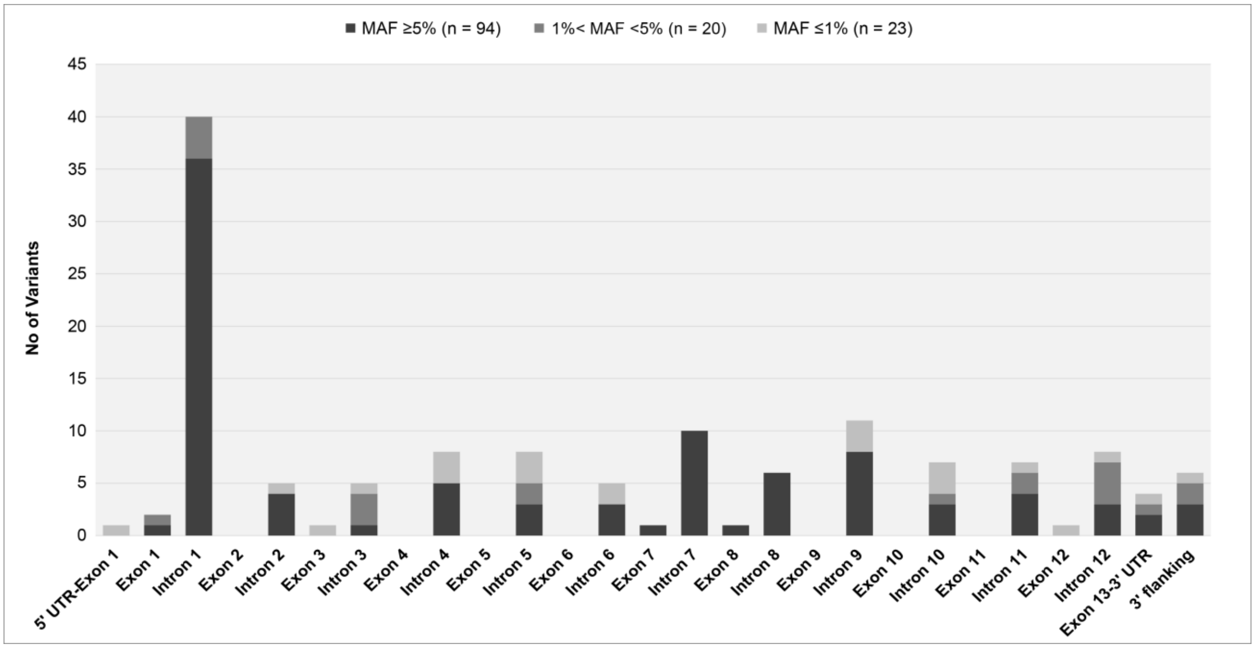

**Figure B5. Regional and minor allele frequency (MAF) distributions of 137 *SCARB1* genotyped variants.**

Details for each variant are shown in **Table B6**. The *SCARB1* RefSeq (hg19, NM_005505) was obtained from CHIP Bioinformatics. UTR, untranslated region.