APPENDIX A

SUPPLEMENTAL MATERIAL FOR CHAPTER 6.0. GENETIC INFLUENCE OF *SCARB1* VARIANTS ON LIPID TRAITS IN US NON-HISPANIC WHITES

Impact of Genetic Variants in Human Scavenger Receptor Class B Type I (*SCARB1*) on Plasma Lipid Traits

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Table A1. Characteristics and lipid profile of the entire NHW sample.

Variables	NHW Samples* (n = 623)
Age, years	52.83 ± 11.41
Body Mass Index, kg/m^2	25.51 ± 4.06
Total Cholesterol, mg/dL	216.99 ± 43.55
LDL-Cholesterol, mg/dL	136.99 ± 40.80
HDL-Cholesterol, mg/dL	50.76 ± 14.35
Triglycerides, mg/dL)	142.72 ± 93.49
Apolipoprotein A1†, mg/dL	149.62 ± 33.33
Apolipoprotein B†, mg/dL	87.72 ± 24.27

HDL-Cholesterol, high density lipoprotein cholesterol; LDL-Cholesterol, low-density lipoprotein cholesterol; NHW, Non-Hispanic White.
Values are presented as mean ± standard deviation (SD), otherwise mentioned.
All data were unadjusted and not excluded the outliers (values beyond means ± 3.5 SD).
*Including individuals with missing data or outliers.
†Measured in a subset of 425 NHW individuals.

Table A2. Characteristics and lipid profile of 95 NHWs with extreme* HDL-C levels.

Variables	High HDL-C Group (HDL-C range†: 58-106 mg/dL)	Low HDL-C Group (HDL-C range†: 20-40 mg/dL)	*P*‡
N (Females, %)	47 (48.9)	48 (50.0)	
Age, years	55.45 ± 9.80	53.03 ± 10.54	0.25
Body Mass Index, kg/m^2	23.17 ± 3.17	27.35 ± 3.90	1.20E-07
Total Cholesterol, mg/dL	227.34 ± 51.76	208.81 ± 44.65	0.07
LDL-Cholesterol, mg/dL	126.84 ± 46.95	125.54 ± 54.97	0.90
HDL-Cholesterol, mg/dL	77.68 ± 13.32	31.81 ± 4.37	2.20E-16
Triglycerides, mg/dL	114.09 ± 60.88	240.21 ± 153.22	1.70E-06

HDL-C or HDL-Cholesterol, high density lipoprotein cholesterol; LDL-Cholesterol, low-density lipoprotein cholesterol; NHW, Non-Hispanic White.
Values are presented as unadjusted means ± standard deviation, otherwise mentioned.
*Adjusted HDL-C distribution for sex and age: HDL-C levels ≥90th %tile, defined as the "High HDL-C group"; HDL-C levels ≤10th %tile, defined as the "Low HDL-C group".
†Unadjusted values.
‡Unadjusted *P*-values were calculated with t-test or χ^2 test depending on types of variables.

Table A3. Primer sequences and sizes of 14 polymerase chain reaction (PCR) fragments, and sizes of 13 *SCARB1* exons based on the *SCARB1* RefSeq* (hg19, NM_005505).

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	

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

Table A4. List of 35 *SCARB1* common HapMap-CEU tagSNPs (HapMap release #27, genome build 36, dbSNP build 126) identified by Haploview Tagger analysis at r^2 ≥0.8, which captured a total of 81 common HapMap-CEU tagSNPs (MAF ≥5%) with a mean r^2 of 0.97 (see linkage disequilibrium structure of 81 *SCARB1* common HapMap-CEU tagSNPs in Figure A1).

Bin #	TagSNP	Alleles Captured
1	rs838912	rs838909, rs838913, rs12819677, rs5888, rs865716, rs3782287, rs989892, rs10773105
2	rs7967406	rs7135223, rs12310272, rs4765616, rs11057838, rs1902569, rs4765618, rs4765178
3	rs838893	rs838891, rs838894, rs838887, rs838896, rs838892
4	**rs838880***	rs838884, rs838882, rs838881, rs838879
5	rs10846749	rs4765623, rs10846748, rs4765621, rs10846751
6	rs4765622	rs12582221, rs4765624, rs4765180
7	rs3924313	rs7137797, rs7306660
8	rs10846745	rs7967521, rs11608336
9	rs11057830	rs10846744, rs11057841
10	rs9919713	rs838867, rs838865
11	rs7134858	rs838900
12	rs2343394*	rs745529
13	rs11057844*	rs7954519
14	rs10773111	rs7954697
15	rs12229555	rs10846753
16	rs11615630	rs12831105
17	rs11057864	rs11057852
18	rs12370382	
19	rs11057869	
20	rs11057851	
21	rs12581963	
22	**rs838883**	
23	rs7139401	
24	rs838905	
25	rs11057820*	
26	**rs4765615***	
27	rs4765181	
28	rs10773107	
29	rs12580803	
30	rs1031605	
31	rs11057853	
32	rs701106*	
33	rs7977729	
34	rs10744182	
35	rs10773109	

CEU, Utah residents with northern and western European ancestry; SNP, single nucleotide polymorphism.
Of total 35 common HapMap-CEU tagSNPs, three were already chosen for genotyping (shown in **bold**), and 32 were selected for genotyping in addition to 40 selected sequence variants.
*HapMap-CEU tagSNPs with significant evidence of single-site association (P <0.05; Tables A8-A11) with any of the 4 lipid traits in this study.

Table A5. Characteristics of 80 *SCARB1* identified variants (44 sequence variants, 32 common HapMap-CEU tagSNPs, and 4 additional relevant variants from the literatures).

					Genotyped Variants (n = 69) in 623 NHW Subjects					Sequence Variants (n = 44)* in 95 NHWs with Extreme HDL-C Levels		
SNP Name†	SNP ID‡	Chr12 Position†	Location	Amino Acid Change	HWE *P*	%Call Rate	Alleles	MA	MAF	MAF in High HDL-C Group	MAF in Low HDL-C Group	Source of Variants
p1257	rs4238001	125348263	Exon 1	Gly2Ser	0.7811	99	G/A	A	0.0818	0.177	0.052	SEQ
p1588		125347932	Intron 1		1	98.6	G/C	C	0.0008	**0.011**	0	SEQ
p4072	rs7139401	125345448	Intron 1		0.2778	99.4	C/T	T	0.4564			HapMap
p5055	rs11057869	125344465	Intron 1		1	99	G/A	A	0.0875			HapMap
p7650	rs11615630	125341870	Intron 1		0.328	99	G/A	A	0.3817			HapMap
p10292	rs4765181	125339228	Intron 1		0.6261	99.2	G/T	T	0.3972			HapMap
p13570	rs11057864	125335950	Intron 1		0.6277	99.7	G/T	T	0.0427			HapMap
p16565	rs10773111	125332955	Intron 1		0.8542	99.4	G/A	A	0.4249			HapMap
p20207	rs11057853	125329313	Intron 1		0.661	98.9	A/G	G	0.4269			HapMap
p20741	rs11057851	125328779	Intron 1		0.8818	99.2	C/T	T	0.1157			HapMap
p21145	rs3924313	125328375	Intron 1		0.2476	99.2	C/T	T	0.3083			HapMap
p22116	rs12370382	125327404	Intron 1		0.3567	99.2	G/A	A	0.4005			HapMap
p26525	rs10773109	125322995	Intron 1		0.3783	99.4	C/G	G	0.4176			HapMap
p28137	rs12229555	125321383	Intron 1		0.796	99	A/G	G	0.2188			HapMap
p28692	rs4765622	125320828	Intron 1		1	99.2	C/T	T	0.4693			HapMap
p28957	rs11057844	125320563	Intron 1		0.655	99.5	G/A	A	0.1839			HapMap
p31072	rs10846749	125318448	Intron 1		0.6064	98.2	C/G	G	0.3448			HapMap
p31938	rs10744182	125317582	Intron 1		0.1471	98.7	A/G	G	0.4130			HapMap
p32129	rs10773107	125317391	Intron 1		0.1424	98.2	G/T	T	0.4828			HapMap
p32273	rs12580803	125317247	Intron 1		0.62	99	T/C	C	0.1726			HapMap
p32395	rs12581963	125317125	Intron 1		0.8394	99.7	C/T	T	0.0829			HapMap
p32860	rs7967406	125316660	Intron 1		0.7176	99.7	A/C	C	0.3921			HapMap
p36908	rs10846745	125312612	Intron 1		0.9054	98.9	C/G	G	0.4748			HapMap
p37095	rs10846744	125312425	Intron 1		0.9472	98.9	C/G	G	0.1364			Addition
p42467	rs11057830	125307053	Intron 1		0.7943	99	C/T	T	0.1329			HapMap
p47235	rs199779577	125302285	Intron 1		1	99	C/T	T	0.0016	**0.011**	0	SEQ
p47416	rs73227571	125302104	Exon 2	Tyr92Tyr	1	100	C/T	T	0.0008	**0.011**	0	SEQ
p48969	rs2343394	125300551	Intron 2		0.3487	98.6	C/T	T	0.2850			HapMap
p49518	rs144194221	125300002	Intron 2		1	99.8	G/A	A	0.0217	0.021	0.031	SEQ
p49690	rs4765615	125299830	Intron 2		0.5216	98.9	A/G	G	0.4497	0.479	0.510	SEQ
p49978	rs5891	125299542	Exon 3	Val135Ile	1	100	G/A	A	0.0096	**0.021**	0	SEQ
p50151	rs2278986	125299369	Intron 3		0.4123	98.9	T/C	C	0.2890	0.326	0.354	SEQ
p50432		125299088	Intron 3		1	99.7	C/T	T	0.0040	**0.011**	0	SEQ
p52556	rs11057820	125296964	Intron 4		0.158	99.4	A/G	G	0.4871			HapMap
p53093	rs201977189	125296427	Exon 5	Gly239Arg	1	99	G/A	A	0.0016	**0.021**	0	SEQ
p53158§	rs61932577	125296362	Intron 5		1	97.9	C/T	T	0.091	0.076	0.106	SEQ
p53248	rs140613481	125296272	Intron 5		1	98.6	T/A	A	0.0008	0	**0.010**	SEQ
p53440§	rs55834271	125296080	Intron 5		1	97.9	T/C	C	0.269	0.293	0.245	SEQ
p53490§		125296030	Intron 5		1	97.9	C/A	A	0.005	0	**0.011**	SEQ
p54492	rs61762481	125295028	Intron 5		0.2869	99.4	G/A	A	0.0468	0.043	0.042	SEQ
p54866		125294654	Intron 6		1	99.4	G/A	A	0.0008	**0.011**	0	SEQ
p55963	rs7134858	125293557	Intron 6		0.1059	99.4	C/T	T	0.0759			HapMap
p56845	rs838902	125292675	Intron 6		1	98.7	A/G	G	0.0211	0.011	0.021	SEQ
p57308		125292212	Intron 7		1	99	C/T	T	0.0016	0	**0.010**	SEQ
p57508	rs71458866	125292012	Intron 7		1	98.9	G/A	A	0.0162	0.011	0.021	SEQ
p57592	rs838903	125291928	Intron 7		1	99.5	G/A	A	0.0161	0.011	0.021	SEQ
p57618		125291902	Intron 7		1	99.2	G/A	A	0.0008	**0.011**	0	SEQ
p58514	rs838905	125291006	Intron 7		0.9407	99.5	T/C	C	0.0347			HapMap
p60255	rs3782287	125289265	Intron 7		0.3306	99.4	C/T	T	0.4766			Addition
p63483	rs838912	125286037	Intron 7		0.287	98.6	G/A	A	0.4927			HapMap
p64285		125285235	Intron 7		1	98.9	C/G	G	0.0008	0	**0.010**	SEQ

					Genotyped Variants (n = 69) in 623 NHW Subjects					Sequence Variants (n = 44)* in 95 NHWs with Extreme HDL-C Levels		
SNP Name†	SNP ID‡	Chr12 Position†	Location	Amino Acid Change	HWE *P*	%Call Rate	Alleles	MA	MAF	MAF in High HDL-C Group	MAF in Low HDL-C Group	Source of Variants
p64772	rs5888	125284748	Exon 8	Ala350Ala	0.3984	98.7	C/T	T	0.4846	0.489	0.490	SEQ
p69612	rs147238482	125279908	Intron 8		1	100	G/A	A	0.0048	0	**0.010**	SEQ
p70038 del5§ [70038_ 70042]		125279482	Intron 9		1	97.9	[GTTTT /-]	del GT TT T	0.005	0	**0.010**	SEQ
p70088§	rs117585141	125279432	Intron 9		1	97.9	G/A	A	0.005	**0.011**	0	SEQ
p70095§		125279425	Intron 9		1	97.9	T/C	C	0.005	**0.011**	0	SEQ
p70292§	rs150388176	125279228	Intron 9		1	96.8	C/T	T	0.005	**0.011**	0	SEQ
p72551	rs1031605	125276969	Intron 9		0.8918	99.7	C/T	T	0.1755			HapMap
p76757	rs9919713	125272763	Intron 9		0.4306	99.5	A/T	T	0.0379			HapMap
p77250	rs201901986	125272270	Intron 9		1	99.8	G/T	T	0.0225	0.043	0.042	SEQ
p77251	rs34339961	125272269	Intron 9		1	99.5	A/T	T	0.0226	0.043	0.042	SEQ
p77842	rs2272310	125271678	Intron 10		0.2367	98.9	G/A	A	0.0146	**0.011**	0	SEQ
p78334		125271186	Intron 10		1	100	T/G	G	0.0024	0	**0.011**	SEQ
p78430	rs838897	125271090	Intron 10		0.3776	99.2	C/G	G	0.0364	0.053	0.031	SEQ
p78747	rs2293440	125270773	Intron 11		0.6055	99.2	T/C	C	0.0243	0.043	0.031	SEQ
p79721	rs838896	125269799	Intron 11		0.3849	99	G/C	C	0.3169			Addition
p79828	rs838895	125269692	Intron 11		0.5691	97.4	C/G	G	0.2891			Addition
p80045	rs838893	125269475	Intron 11		0.808	99.5	G/A	G	0.2879			HapMap
p83088	rs7977729	125266432	Intron 12		1	99.5	A/G	G	0.2669			HapMap
p83884	rs701106	125265636	Intron 12		0.514	99.8	C/T	T	0.1527			HapMap
p86316‖	rs701104	125263204	Intron 12		0.527	100	G/T	T	0.105	0.138	0.073	SEQ
p86436	rs10396214	125263084	Exon 13-3' UTR	Arg484Trp#	1	100	C/T	T	0.0088	0.021	0.010	SEQ
p87004	rs184715678	125262516	Exon 13-3' UTR		1	99.7	C/A	A	0.0016	0	**0.010**	SEQ
p87210		125262310	Exon 13-3' UTR		1	100	G/A	A	0.0016	0	**0.010**	SEQ
p87381	rs192190977	125262139	3' flanking		1	99.7	C/T	T	0.0024	**0.011**	0	SEQ
p87416‖	rs838884	125262104	3' flanking		0.6666	98.9	T/C	C	0.335	0.402	0.271	SEQ
p87681	rs838883	125261839	3' flanking		0.3215	99.7	G/A	A	0.0845	0.138	0.073	SEQ
p87707‖	rs838882	125261813	3' flanking		0.5149	97.9	A/G	G	0.328	0.389	0.271	SEQ
p87723‖	rs838881	125261797	3' flanking		0.4529	98.9	T/C	C	0.330	0.391	0.271	SEQ
p87927	rs838880	125261593	3' flanking		0.6556	99.7	A/G	G	0.3237	0.415	0.281	SEQ

Addition, additional relevant variants from the literatures; CEU, Utah residents with northern and western European ancestry; del, deletion; HWE, Hardy-Weinberg equilibrium; HDL-C, high-density lipoprotein cholesterol; MA, minor allele; MAF, minor allele frequency; NHW, Non-Hispanic White; SEQ, sequencing; SNP, single nucleotide polymorphism; UTR, untranslated region.

All alleles on reverse strand.

A list of 32 common HapMap-CEU tagSNPs is shown in Table A4.

Grey highlighted cells present data derived from sequencing data due to not chosen for genotyping or genotyping failure.

The MAFs of 44 sequence variants specific to one of the two extreme HDL-C groups are shown in **bold**.

*A total of 44 sequence variants were comprised of one 5-bp deletion and 43 substitutional variants, which included 33 transition (A ↔ G, n = 18; C ↔ T, n = 15), and 10 transversion (A ↔ C, n = 2; A ↔ T, n = 2; C ↔ G, n = 3; G ↔ T, n = 3).

A total of 69 genotyped variants (33 variants identified by sequencing, 32 common HapMap-CEU tagSNPs, and 4 additional relevant variants from the literatures) that passed genotyping quality controls were included in downstream analyses.

A linkage disequilibrium structure of 69 genotyped variants that passed genotyping quality controls and were included in downstream analyses is shown in Figure A3.

†Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.

‡dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).

§Sequence variants but failed genotyping (n = 7).

||Sequence variants (n = 4) that not chosen for genotyping due to high linkage disequilibrium (r^2 ≥0.90; Figure A2): p86316 were tagged with p87681; p87416, p87707, p87723 were tagged with p87927.

#rs10396214 is translated leading to amino acid change (Arg484Trp) in isoform 2 (alternative splice form); not translated in isoform 1.

Table A6. Distribution of 69 *SCARB1* genotyped variants.

	Total N (%)	MAF ≥5% n (%)	MAF between 1-5% n (%)	MAF ≤1% n (%)
Total Variants	**69 (100.00)**	**39 (56.52)**	**13 (18.84)**	**17 (24.64)**
By Identified/Novel*				
Identified	61 (88.40)	39 (56.52)	13 (18.84)	9 (13.04)
Novel*	8 (11.60)			8 (11.60)
By Locations				
Exons	5 (7.25)	2 (2.90)		3 (4.35)
Introns	58 (84.05)	35 (50.72)	13 (18.84)	10 (14.49)
Exon-3' UTR	3 (4.35)			3 (4.35)
3' flanking	3 (4.35)	2 (2.90)		1 (1.45)
By Amino Acid Changes				
Non-synonymous†	4†	1		3†
Synonymous	2	1		1

MAF, minor allele frequency; UTR, untranslated region.
*dbSNP build 139: GRCh37.p10; novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).
†Including rs10396214 (Arg484Trp; MAF = 0.0088) located in exon 13-3' UTR, which is translated in isoform 2.

Table A7. Characteristics of 11 *SCARB1* novel* variants.

SNP Name†	Chr12 Position†	Location	RegulomeDB Score‡	Major/Minor Alleles	MAF	MA Effect on Trait§
p1588	125347932	Intron 1	4	G/C	0.0008	
p50432	125299088	Intron 3	2b	C/T	0.004	
p53490	125296030	Intron 5	7	C/A	0.005‖	
p54866	125294654	Intron 6	4	G/A	0.0008	↑ HDL-C
p57308	125292212	Intron 7	4	C/T	0.0016	↓ TG
p57618	125291902	Intron 7	4	G/A	0.0008	↑ HDL-C
p64285	125285235	Intron 7	3a	C/G	0.0008	↑ TG
p70038del5 [70038_70042]	125279482	Intron 9	6	[GTTTT/-]	0.005‖	
p70095	125279425	Intron 9	7	T/C	0.005‖	
p78334	125271186	Intron 10	4	T/G	0.0024	↑ ApoB
p87210	125262310	Exon 13-3' UTR	2b	G/A	0.0016	

ApoB, apolipoprotein B; del, deletion; HDL-C, high-density lipoprotein cholesterol; MA, minor allele; MAF, minor allele frequency; SNP, single nucleotide polymorphism; TG, triglycerides; UTR, untranslated region; ↑, increased; ↓, decreased.
All alleles on reverse strand.
*dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).
†Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
‡The RegulomeDB (version 1.0) scoring scheme represented as following: **score 1a**, eQTL, 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.
§Allele effects are based on single-site association results, see details in Tables 6.4, A8, A9, and A11.
‖Derived from sequencing data due to genotyping failure.

Table A8. Single-site association results of 69 *SCARB1* genotyped variants for high-density lipoprotein cholesterol (HDL-C).

SNP Name*	SNP ID†	Chr12 Position*	Location	Amino Acid Change	RegDB Score‡	MA, MAF	Genotype	Genotype Count	Adjusted Mean (mg/dL)	SD (mg/dL)	β	SE	*P*	FDR§
SNPs with MAF ≥5%														
p1257	rs4238001	125348263	Exon 1	Gly2Ser	4	A, 0.0818	AA/GA/GG	5/91/519	48.34/52.42/50.24	12.70/15.14/13.86	0.0224	0.0193	0.2455	0.632
p4072	rs7139401	125345448	Intron 1		3b	T, 0.4564	TT/CT/CC	135/293/189	51.69/50.42/50.03	14.92/14.00/13.50	0.0097	0.0105	0.3552	0.682
p5055	rs11057869	125344465	Intron 1		7	A, 0.0875	GG/GA/AA	512/98/5	50.19/52.48/48.34	13.74/15.61/12.70	0.0220	0.0188	0.2433	0.632
p7650	rs11615630	125341870	Intron 1		5	A, 0.3817	GG/GA/AA	241/279/95	50.15/51.41/49.72	13.93/14.18/13.81	0.0006	0.0108	0.9545	0.954
p10292	rs4765181	125339228	Intron 1		5	T, 0.3972	TT/GT/GG	93/303/220	50.53/50.09/51.26	15.14/13.06/14.84	-0.0074	0.0110	0.5047	0.696
p16565	rs10773111	125332955	Intron 1		6	A, 0.4249	GG/GA/AA	202/306/109	50.77/50.52/50.31	14.96/13.36/14.16	-0.0011	0.0108	0.9160	0.933
p20207	rs11057853	125329313	Intron 1		5	G, 0.4269	GG/AG/AA	109/307/198	49.73/51.02/50.47	14.88/13.78/14.08	-0.0047	0.0109	0.6641	0.804
p20741	rs11057851	125328779	Intron 1		5	T, 0.1157	TT/CT/CC	9/125/482	45.25/51.69/50.45	7.11/14.02/14.14	0.0073	0.0166	0.6597	0.804
p21145	rs3924313	125328375	Intron 1		6	T, 0.3083	TT/CT/CC	52/275/289	50.2/50.47/50.96	14.41/14.76/13.26	-0.0101	0.0118	0.3898	0.682
p22116	rs12370382	125327404	Intron 1		1f	A, 0.4005	GG/GA/AA	215/308/93	50.99/50.18/50.53	14.70/13.61/13.53	-0.0031	0.0110	0.7775	0.894
p26525	rs10773109	125322995	Intron 1		4	G, 0.4176	GG/CG/CC	102/312/203	49.46/50.42/51.26	13.18/13.90/14.38	-0.0132	0.0109	0.2286	0.632
p28137	rs12229555	125321383	Intron 1		7	G, 0.2188	GG/AG/AA	31/207/377	49.96/51.16/50.23	12.18/14.49/13.95	0.0080	0.0128	0.5310	0.705
p28692	rs4765622	125320828	Intron 1		5	T, 0.4693	TT/CT/CC	136/307/173	51.64/50.17/50.23	13.82/13.74/14.79	0.0130	0.0107	0.2255	0.632
p28957	rs11057844	125320563	Intron 1		5	A, 0.1839	GG/GA/AA	413/182/23	51.41/49.51/45.02	14.05/14.29/9.08	-0.0395	0.0135	**0.0035**	0.227
p31072	rs10846749	125318448	Intron 1		4	G, 0.3448	CC/CG/GG	266/269/75	50.28/50.7/51.23	13.43/14.53/14.23	0.0059	0.0111	0.5976	0.767
p31938	rs10744182	125317582	Intron 1		5	G, 0.4130	GG/AC/AA	114/280/219	50.31/50.32/51.31	14.04/14.05/14.08	-0.0088	0.0106	0.4065	0.682
p32129	rs10773107	125317391	Intron 1		7	T, 0.4828	TT/GT/GG	132/324/154	48.85/51.05/50.98	13.52/13.80/15.13	-0.0140	0.0111	0.2065	0.632
p32273	rs12580803	125317247	Intron 1		5	C, 0.1726	TT/TC/CC	418/181/16	50.51/50.27/54.34	13.88/14.40/15.90	0.0045	0.0144	0.7527	0.880
p32395	rs12581963	125317125	Intron 1		5	T, 0.0829	TT/CT/CC	5/93/521	54.78/51.09/50.5	23.81/14.00/13.95	0.0129	0.0191	0.4973	0.696
p32860	rs7967406	125316660	Intron 1		6	C, 0.3921	CC/AC/AA	96/291/232	51.27/50.25/50.64	13.90/14.29/13.86	0.0023	0.0109	0.8303	0.933
p36908	rs10846745	125312612	Intron 1		4	G, 0.4748	CC/CG/GG	169/305/140	50.82/49.87/51.62	14.34/13.79/14.45	0.0051	0.0107	0.6330	0.794
p37095	rs10846744	125312425	Intron 1		4	G, 0.1364	CC/CG/GG	458/144/12	50.92/49.43/52.28	14.20/13.55/16.60	-0.0132	0.0155	0.3943	0.682
p42467	rs11057830	125307053	Intron 1		7	T, 0.1329	TT/CT/CC	12/140/463	55.56/48.99/51.06	18.40/13.04/14.23	-0.0120	0.0156	0.4429	0.682
p48969	rs2343394	125300551	Intron 2		5	T, 0.2850	TT/CT/CC	53/240/319	49.8/50.09/50.93	13.02/14.11/14.27	-0.0090	0.0117	0.4412	0.682
p49690	rs4765615	125299830	Intron 2		5	G, 0.4497	GG/AG/AA	118/314/182	49.97/50.26/51.6	13.98/13.87/14.54	-0.0133	0.0110	0.2263	0.632

p50151	rs2278986	125299369	Intron 3		5	C, 0.2890	CC/TC/TT	54/244/316	50.29/50.29/50.72	12.85/14.42/13.99	-0.0040	0.0117	0.7308	0.869
p52556	rs11057820	125296964	Intron 4		5	G, 0.4871	GG/AG/AA	154/291/172	50.53/50.22/51.24	13.83/14.23/13.99	-0.0054	0.0104	0.6003	0.767
p55963	rs7134858	125293557	Intron 6		6	T, 0.0759	TT/CT/CC	6/80/531	48.96/48.91/50.92	11.83/12.75/14.25	-0.0235	0.0196	0.2305	0.632
p60255	rs3782287	125289265	Intron 7		5	T, 0.4766	CC/CT/TT	174/296/147	51.9/50.07/50.13	14.98/13.99/13.05	-0.0122	0.0104	0.2448	0.632
p63483	rs838912	125286037	Intron 7		7	A, 0.4927	AA/GA/GG	154/293/165	52.26/49.91/50.36	14.88/14.02/13.31	0.0125	0.0104	0.2308	0.632
p64772	rs5888	125284748	Exon 8	Ala350Ala	3a	T, 0.4846	CC/CT/TT	169/296/148	50.62/49.9/51.81	13.27/14.22/14.65	0.0068	0.0106	0.5213	0.705
p72551	rs1031605	125276969	Intron 9		5	T, 0.1755	CC/CT/TT	419/182/18	50.16/51.32/54.26	14.08/14.04/13.00	0.0252	0.0141	0.0731	0.586
p79721	rs838896	125269799	Intron 11		5	C, 0.3169	GG/GC/CC	292/257/66	49.98/51.06/51.05	13.64/14.12/15.75	0.0086	0.0114	0.4506	0.682
p79828	rs838895	125269692	Intron 11		5	G, 0.2891	GG/CG/CC	53/243/309	51.83/51.4/49.86	16.27/13.97/13.64	0.0169	0.0117	0.1489	0.632
p80045	rs838893	125269475	Intron 11		5	A, 0.2879	GG/GA/AA	315/251/52	49.92/51.03/51.94	13.63/14.11/16.24	0.0136	0.0117	0.2472	0.632
p83088	rs7977729	125266432	Intron 12		6	G, 0.2669	GG/AG/AA	44/242/332	49.62/51.6/49.9	12.79/14.60/13.73	0.0107	0.0121	0.3768	0.682
p83884	rs701106	125265636	Intron 12		5	T, 0.1527	TT/CT/CC	17/155/448	53.12/52.62/49.76	16.14/14.18/13.88	0.0394	0.0144	**0.0066**	0.227
p87681	rs838883	125261839	3' flanking		5	A, 0.0845	GG/GA/AA	518/99/2	50.4/51.3/57.57	13.89/14.31/38.18	0.0152	0.0197	0.4416	0.682
p87927	rs838880	125261593	3' flanking		5	G, 0.3237	AA/AG/GG	281/276/62	49.11/51.85/51.34	13.27/14.45/15.33	0.0257	0.0114	**0.0250**	0.457
SNPs with MAF between 1-5%														
p13570	rs11057864	125335950	Intron 1		4	T, 0.0427	GT/GG	53/566	52.64/50.43	16.71/13.76	0.0236	0.0268	0.3795	0.682
p49518	rs144194221	125300002	Intron 2		5	A, 0.0217	GA/GG	27/593	50.92/50.56	14.32/14.03	0.0046	0.0370	0.9017	0.933
p54492	rs61762481	125295028	Intron 5		4	A, 0.0468	AA/GA/GG	3/52/562	43.03/51.6/50.53	1.73/14.25/14.07	0.0034	0.0245	0.8882	0.933
p56845	rs838902	125292675	Intron 6		5	G, 0.0211	AA/AG	587/26	50.55/51.28	13.91/16.20	0.0059	0.0373	0.8737	0.933
p57508	rs71458866	125292012	Intron 7		4	A, 0.0162	GA/GG	20/594	50.91/50.52	17.53/13.96	-0.0043	0.0426	0.9191	0.933
p57592	rs838903	125291928	Intron 7		4	A, 0.0161	GA/GG	20/598	50.92/50.53	17.53/13.95	-0.0045	0.0425	0.9163	0.933
p58514	rs838905	125291006	Intron 7		4	C, 0.0347	TC/TT	43/575	53.28/50.34	17.13/13.80	0.0372	0.0296	0.2089	0.632
p76757	rs9919713	125272763	Intron 9		6	T, 0.0379	AA/AT/TT	573/43/2	50.33/52.93/58.63	13.76/17.42/25.46	0.0339	0.0272	0.2127	0.632
p77250	rs201901986	125272270	Intron 9		6	T, 0.0225	GG/GT	592/28	50.47/53.14	13.87/17.63	0.0297	0.0362	0.4112	0.682
p77251	rs34339961	125272269	Intron 9		6	T, 0.0226	AA/AT	590/28	50.45/53.11	13.88/17.63	0.0296	0.0362	0.4144	0.682
p77842	rs2272310	125271678	Intron 10		5	A, 0.0146	AA/GA/GG	1/16/597	48.44/54.85/50.41	NA/18.25/13.93	0.0414	0.0424	0.3296	0.682
p78430	rs838897	125271090	Intron 10		5	G, 0.0364	GG/CG/CC	2/41/573	58.69/53.16/50.42	25.46/17.63/13.75	0.0355	0.0276	0.1996	0.632
p78747	rs2293440	125270773	Intron 11		5	C, 0.0243	CC/TC/TT	1/28/587	48.54/54.01/50.45	NA/17.43/13.90	0.0378	0.0339	0.2647	0.652
SNPs with MAF ≤1%														
p1588		125347932	Intron 1		4	C, 0.0008	GC/GG	1/611	80.6/50.55	NA/13.98	0.3089	0.1879	0.1008	0.586
p47235	rs199779577	125302285	Intron 1		4	T, 0.0016	CC/CT	613/2	50.51/59.91	14.06/25.46	0.1240	0.1331	0.3520	0.682

p47416	rs73227571	125302104	Exon 2	Tyr92Tyr	5	T, 0.0008	CC/CT	620/1	50.55/67.52	14.03/NA	0.2718	0.1878	0.1484	0.632
p49978	rs5891	125299542	Exon 3	Val135Ile	5	A, 0.0096	GA/GG	12/609	53.89/50.52	16.92/13.98	0.0454	0.0544	0.4043	0.682
p50432		125299088	Intron 3		2b	T, 0.0040	CC/CT	614/5	50.52/55.26	14.06/15.12	0.0745	0.0841	0.3762	0.682
p53093	rs201977189	125296427	Exon 5	Gly239Arg	5	A, 0.0016	GA/GG	2/613	71.1/50.51	10.61/14.02	0.2937	0.1329	**0.0275**	0.457
p53248	rs140613481	125296272	Intron 5		5	A, 0.0008	TA/TT	1/611	35.43/50.59	NA/14.07	-0.3175	0.1877	0.0913	0.586
p54866		125294654	Intron 6		4	A, 0.0008	GA/GG	1/616	87.87/50.52	NA/13.95	0.3998	0.1873	**0.0332**	0.457
p57308		125292212	Intron 7		4	T, 0.0016	CC/CT	613/2	50.56/44.58	14.08/12.02	-0.0994	0.1329	0.4547	0.682
p57618		125291902	Intron 7		4	A, 0.0008	GA/GG	1/615	86.08/50.54	NA/13.96	0.3727	0.1873	**0.0470**	0.517
p64285		125285235	Intron 7		3a	G, 0.0008	CC/CG	613/1	50.59/34.34	14.05/NA	-0.3643	0.1875	0.0525	0.517
p69612	rs147238482	125279908	Intron 8		4	A, 0.0048	GA/GG	6/615	49.52/50.59	13.72/14.05	-0.0154	0.0767	0.8408	0.933
p78334		125271186	Intron 10		4	G, 0.0024	TG/TT	3/618	39.28/50.64	11.02/14.04	-0.1888	0.1080	0.0811	0.586
p86436	rs10396214	125263084	Exon 13-3' UTR	Arg484Trp[#]	4	T, 0.0088	CC/CT	610/11	50.62/48.33	14.04/14.46	-0.0402	0.0570	0.4808	0.696
p87004	rs184715678	125262516	Exon 13-3' UTR		2b	A, 0.0016	CA/CC	2/617	39.02/50.59	11.31/14.05	-0.2166	0.1323	0.1019	0.586
p87210		125262310	Exon 13-3' UTR		2b	A, 0.0016	GG/GA	619/2	50.6/46.35	14.03/16.97	-0.0914	0.1326	0.4911	0.696
p87381	rs192190977	125262139	3' flanking		2b	T, 0.0024	CC/CT	616/3	50.53/55.46	14.07/14.19	0.0885	0.1085	0.4149	0.682

FDR, false discovery rate; MA, minor allele; MAF, minor allele frequency; NA, not analyzed; RegDB, RegulomeDB; SD, standard deviation; SE, standard error; SNP, single nucleotide polymorphism; UTR, untranslated region.
All alleles on reverse strand. HDL-C values were Box-Cox transformed . Results were adjusted for covariates: sex, age, smoking, and body mass index.
Significant *P*-values (*P* <0.05) are shown in **bold**.
*Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
†dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).
‡Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table A7 or can be seen at http://regulome.stanford.edu/help.
§FDR is referred to *q*-value in Benjamini-Hochberg procedure.
#rs10396214 is translated leading to amino acid change (Arg484Trp) in isoform 2 (alternative splice form); not translated in isoform 1.

Table A9. Single-site association results of 69 *SCARB1* genotyped variants for apolipoprotein B (ApoB).

SNP Name*	SNP ID†	Chr12 Position*	Location	Amino Acid Change	RegDB Score‡	MA, MAF	Genotype	Genotype Count	Adjusted Mean (mg/dL)	SD (mg/dL)	β	SE	*P*	FDR§
SNPs with MAF ≥5%														
p1257	rs4238001	125348263	Exon 1	Gly2Ser	4	A, 0.0818	AA/GA/GG	5/64/360	82.7/89.62/87.61	21.03/22.19/24.03	0.2786	0.7655	0.7161	0.919
p4072	rs7139401	125345448	Intron 1		3b	T, 0.4564	TT/CT/CC	91/199/142	88.38/89.21/85.58	21.82/24.86/23.87	0.4818	0.4323	0.2657	0.583
p5055	rs11057869	125344465	Intron 1		7	A, 0.0875	GG/GA/AA	353/71/5	87.73/88.83/82.7	24.14/21.85/21.03	0.1155	0.7433	0.8766	0.978
p7650	rs11615630	125341870	Intron 1		5	A, 0.3817	GG/GA/AA	181/192/59	85.26/89.94/89.15	22.75/25.10/23.11	0.7301	0.4533	0.1080	0.456
p10292	rs4765181	125339228	Intron 1		5	T, 0.3972	TT/GT/GG	75/211/146	86.99/88.37/87.8	23.79/23.82/24.47	-0.0530	0.4529	0.9069	0.978
p16565	rs10773111	125332955	Intron 1		6	A, 0.4249	GG/GA/AA	139/209/84	91.01/86.26/87.18	21.75/25.06/24.45	-0.6627	0.4432	0.1356	0.488
p20207	rs11057853	125329313	Intron 1		5	G, 0.4269	GG/AG/AA	72/209/149	92.54/87.67/85.99	21.85/25.31/22.41	0.8207	0.4486	0.0680	0.430
p20741	rs11057851	125328779	Intron 1		5	T, 0.1157	TT/CT/CC	7/86/339	94.98/89.57/87.39	23.95/22.31/24.43	0.7897	0.6897	0.2529	0.583
p21145	rs3924313	125328375	Intron 1		6	T, 0.3083	TT/CT/CC	36/184/212	96.17/87.11/87.27	24.25/24.70/23.10	0.6565	0.4912	0.1821	0.507
p22116	rs12370382	125327404	Intron 1		1f	A, 0.4005	GG/GA/AA	143/215/73	88.83/87/89.33	21.78/25.15/24.71	-0.0729	0.4562	0.8730	0.978
p26525	rs10773109	125322995	Intron 1		4	G, 0.4176	GG/CG/CC	67/211/154	88.76/87.41/88.32	23.07/23.45/25.23	0.0192	0.4585	0.9667	0.995
p28137	rs12229555	125321383	Intron 1		7	G, 0.2188	GG/AG/AA	25/135/271	92.49/84.62/89.13	18.76/22.24/25.16	-0.3159	0.5249	0.5476	0.801
p28692	rs4765622	125320828	Intron 1		5	T, 0.4693	TT/CT/CC	109/209/113	89.47/86.41/89.43	27.10/23.04/22.56	-0.0560	0.4406	0.8988	0.978
p28957	rs11057844	125320563	Intron 1		5	A, 0.1839	GG/GA/AA	302/116/14	87.28/90.26/83.11	24.05/23.41/27.36	0.2856	0.5874	0.6271	0.836
p31072	rs10846749	125318448	Intron 1		4	G, 0.3448	CC/CG/GG	189/185/51	88.32/85.7/90.1	24.89/22.79/20.14	0.0045	0.4608	0.9922	0.995
p31938	rs10744182	125317582	Intron 1		5	G, 0.4130	GG/AC/AA	74/201/155	83.5/89.69/87.69	24.46/23.81/24.02	-0.3933	0.4488	0.3813	0.728
p32129	rs10773107	125317391	Intron 1		7	T, 0.4828	TT/GT/GG	97/219/111	89.12/88.28/85.49	22.33/23.80/24.64	0.5435	0.4492	0.2270	0.572
p32273	rs12580803	125317247	Intron 1		5	C, 0.1726	TT/TC/CC	305/111/13	88.13/86.24/95.04	23.87/22.67/28.14	0.0036	0.5974	0.9952	0.995
p32395	rs12581963	125317125	Intron 1		5	T, 0.0829	TT/CT/CC	4/67/362	75.73/86.11/88.42	29.82/19.93/24.57	-0.8103	0.7786	0.2986	0.627
p32860	rs7967406	125316660	Intron 1		6	C, 0.3921	CC/AC/AA	75/206/151	91.23/88.08/85.82	22.56/24.06/24.28	0.7558	0.4475	0.0920	0.447
p36908	rs10846745	125312612	Intron 1		4	G, 0.4748	CC/CG/GG	130/212/87	91.39/86.45/85.98	22.47/23.43/25.64	-0.8517	0.4428	0.0551	0.430
p37095	rs10846744	125312425	Intron 1		4	G, 0.1364	CC/CG/GG	321/98/10	87.89/87.61/90.01	24.46/21.75/26.35	0.0774	0.6321	0.9027	0.978
p42467	rs11057830	125307053	Intron 1		7	T, 0.1329	TT/CT/CC	10/97/323	91.83/87.71/88.03	26.34/21.86/24.46	0.1439	0.6338	0.8205	0.962
p48969	rs2343394	125300551	Intron 2		5	T, 0.2850	TT/CT/CC	41/174/213	95.46/88.93/85.34	26.86/22.18/23.84	1.2544	0.4721	**0.0082**	**0.165**
p49690	rs4765615	125299830	Intron 2		5	G, 0.4497	GG/AG/AA	90/220/120	94/86.95/84.81	23.04/24.06/23.53	1.2493	0.4518	**0.0059**	**0.165**
p50151	rs2278986	125299369	Intron 3		5	C, 0.2890	CC/TC/TT	42/175/211	92.99/89.86/85.07	25.99/23.12/23.65	1.1926	0.4735	**0.0122**	**0.165**
p52556	rs11057820	125296964	Intron 4		5	G, 0.4871	GG/AG/AA	114/203/115	91.57/87.06/85.52	23.64/23.03/25.42	0.8700	0.4300	**0.0436**	0.430

p55963	rs7134858	125293557	Intron 6		6	T, 0.0759	TT/CT/CC	6/61/365	97.9/88.42/87.59	33.43/21.21/24.25	0.6119	0.7659	0.4247	0.738
p60255	rs3782287	125289265	Intron 7		5	T, 0.4766	CC/CT/TT	123/209/100	90.31/86.03/89.07	23.87/24.70/22.36	-0.2158	0.4386	0.6229	0.836
p63483	rs838912	125286037	Intron 7		7	A, 0.4927	AA/GA/GG	111/205/112	91.54/85.35/88.82	23.08/24.45/23.05	0.3773	0.4344	0.3856	0.728
p64772	rs5888	125284748	Exon 8	Ala350Ala	3a	T, 0.4846	CC/CT/TT	117/205/106	89.06/85.2/91.66	23.39/24.51/21.95	0.3416	0.4331	0.4307	0.738
p72551	rs1031605	125276969	Intron 9		5	T, 0.1755	CC/CT/TT	297/122/14	87.21/89.77/87.87	23.45/24.82/27.25	0.4528	0.5830	0.4378	0.738
p79721	rs838896	125269799	Intron 11		5	C, 0.3169	GG/GC/CC	204/173/52	86.22/88.87/90.84	22.17/26.18/20.74	0.6682	0.4561	0.1436	0.488
p79828	rs838895	125269692	Intron 11		5	G, 0.2891	GG/CG/CC	41/170/212	88.68/89.38/86.22	18.51/26.01/22.00	0.5448	0.4724	0.2495	0.583
p80045	rs838893	125269475	Intron 11		5	A, 0.2879	GG/GA/AA	216/175/40	85.98/89.1/91.47	22.05/26.35/21.98	0.7795	0.4799	0.1050	0.456
p83088	rs7977729	125266432	Intron 12		6	G, 0.2669	GG/AG/AA	34/167/230	91.94/89.34/86.32	22.78/26.23/22.36	0.7858	0.4961	0.1140	0.456
p83884	rs701106	125265636	Intron 12		5	T, 0.1527	TT/CT/CC	12/116/305	90.57/85.66/88.56	24.59/22.22/24.52	-0.3865	0.5974	0.5179	0.797
p87681	rs838883	125261839	3' flanking		5	A, 0.0845	GG/GA/AA	361/69/2	88.49/84.39/95.79	24.43/21.12/17.61	-0.8318	0.8089	0.3044	0.627
p87927	rs838880	125261593	3' flanking		5	G, 0.3237	AA/AG/GG	198/189/45	89.56/86.83/84.5	24.46/23.90/21.51	-0.7008	0.4737	0.1398	0.488
SNPs with MAF between 1-5%														
p13570	rs11057864	125335950	Intron 1		4	T, 0.0427	GT/GG	43/390	89.44/87.78	21.71/24.20	0.5236	1.0510	0.6186	0.836
p49518	rs144194221	125300002	Intron 2		5	A, 0.0217	GA/GG	20/413	92.68/87.69	23.99/23.96	1.4134	1.4967	0.3456	0.691
p54492	rs61762481	125295028	Intron 5		4	A, 0.0468	AA/GA/GG	3/39/388	57.72/87.47/88.18	2.17/23.11/23.83	-1.2096	0.9543	0.2056	0.538
p56845	rs838902	125292675	Intron 6		5	G, 0.0211	AA/AG	412/18	88.43/78.59	23.86/26.13	-2.7951	1.5697	0.0757	0.430
p57508	rs71458866	125292012	Intron 7		4	A, 0.0162	GA/GG	14/415	76.85/88.22	21.11/23.69	-3.1506	1.7487	0.0723	0.430
p57592	rs838903	125291928	Intron 7		4	A, 0.0161	GA/GG	14/418	76.87/88.2	22.11/23.93	-3.1378	1.7632	0.0758	0.430
p58514	rs838905	125291006	Intron 7		4	C, 0.0347	TC/TT	30/402	81.02/88.35	25.58/23.79	-2.0859	1.2327	0.0914	0.447
p76757	rs9919713	125272763	Intron 9		6	T, 0.0379	AA/AT/TT	399/31/1	87.92/85.15/104.41	23.64/27.58/NA	-0.4687	1.1423	0.6818	0.892
p77250	rs201901986	125272270	Intron 9		6	T, 0.0225	GG/GT	413/20	88.11/84.59	23.56/31.51	-1.1574	1.4925	0.4385	0.738
p77251	rs34339961	125272269	Intron 9		6	T, 0.0226	AA/AT	412/20	87.99/84.63	23.55/31.51	-1.1124	1.4905	0.4559	0.738
p77842	rs2272310	125271678	Intron 10		5	A, 0.0146	AA/GA/GG	1/11/416	104.05/86.22/87.77	NA/19.84/23.81	0.3875	1.6894	0.8187	0.962
p78430	rs838897	125271090	Intron 10		5	G, 0.0364	GG/CG/CC	1/28/401	104.1/84.26/87.96	NA/28.91/23.59	-0.6962	1.1929	0.5598	0.801
p78747	rs2293440	125270773	Intron 11		5	C, 0.0243	CC/TC/TT	1/18/410	104.03/83.42/88.14	NA/31.99/23.42	-0.8158	1.4170	0.5651	0.801
SNPs with MAF ≤1%														
p1588		125347932	Intron 1		4	C, 0.0008	GC/GG	1/426	57.04/87.78	NA/23.65	-8.6447	6.4463	0.1806	0.507
p47235	rs199779577	125302285	Intron 1		4	T, 0.0016	CC/CT	428/1	87.83/94.39	23.74/NA	2.1039	6.5209	0.7471	0.941
p47416	rs73227571	125302104	Exon 2	Tyr92Tyr	5	T, 0.0008	CC/CT	433/1	88.02/44.8	23.91/NA	-12.3322	6.5369	0.0599	0.430
p49978	rs5891	125299542	Exon 3	Val135Ile	5	A, 0.0096	GA/GG	8/426	109.01/87.52	20.91/23.82	5.8266	2.3105	**0.0120**	**0.165**

p50432		125299088	Intron 3		2b	T, 0.0040	CC/CT	429/3	87.84/88.03	24.01/12.15	0.2876	3.7935	0.9396	0.983
p53093	rs201977189	125296427	Exon 5	Gly239Arg	5	A, 0.0016	GA/GG	2/428	91.85/87.88	1.34/23.87	1.3306	4.6468	0.7748	0.948
p53248	rs140613481	125296272	Intron 5		5	A, 0.0008	TA/TT	1/426	84.58/87.91	NA/23.78	-0.6467	6.5085	0.9209	0.978
p54866		125294654	Intron 6		4	A, 0.0008	GA/GG	1/429	70.04/87.94	NA/23.84	-4.9002	6.5083	0.4519	0.738
p57308		125292212	Intron 7		4	T, 0.0016	CC/CT	428/1	87.89/71.68	23.72/NA	-4.3898	6.4721	0.4980	0.787
p57618		125291902	Intron 7		4	A, 0.0008	GA/GG	NA	NA	NA	NA	NA	NA	NA
p64285		125285235	Intron 7		3a	G, 0.0008	CC/CG	428/1	87.92/58.04	23.69/NA	-8.5632	6.4728	0.1866	0.507
p69612	rs147238482	125279908	Intron 8		4	A, 0.0048	GA/GG	5/429	103.3/87.74	25.94/23.87	4.2242	2.9344	0.1507	0.488
p78334		125271186	Intron 10		4	G, 0.0024	TG/TT	2/432	144.16/87.66	2.05/23.67	14.5804	4.5709	**0.0015**	**0.104**
p86436	rs10396214	125263084	Exon 13-3' UTR	Arg484Trp[#]	4	T, 0.0088	CC/CT	425/9	87.97/85.32	24.05/18.86	-0.6137	2.2065	0.7810	0.948
p87004	rs184715678	125262516	Exon 13-3' UTR		2b	A, 0.0016	CA/CC	2/430	97.77/87.79	17.32/23.98	2.9213	4.6219	0.5277	0.797
p87210		125262310	Exon 13-3' UTR		2b	A, 0.0016	GG/GA	432/2	87.81/111.99	23.85/35.92	6.3910	4.6228	0.1675	0.507
p87381	rs192190977	125262139	3' flanking		2b	T, 0.0024	CC/CT	429/3	87.73/102.92	23.96/19.37	4.2516	3.7760	0.2608	0.583

FDR, false discovery rate; MA, minor allele; MAF, minor allele frequency; NA, not analyzed; RegDB, RegulomeDB; SD, standard deviation; SE, standard error; SNP, single nucleotide polymorphism; UTR, untranslated region.
All alleles on reverse strand. ApoB values were Box-Cox transformed. Results were adjusted for covariates: age and smoking.
Significant *P*-values (*P* <0.05) and FDR values that passed the threshold (FDR <0.20) are shown in **bold**.
*Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
†dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).
‡Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table A7 or can be seen at http://regulome.stanford.edu/help.
§FDR is referred to *q*-value in Benjamini-Hochberg procedure.
#rs10396214 is translated leading to amino acid change (Arg484Trp) in isoform 2 (alternative splice form); not translated in isoform 1.

Table A10. Single-site association results of 69 *SCARB1* genotyped variants for low-density lipoprotein cholesterol (LDL-C).

SNP Name*	SNP ID†	Chr12 Position*	Location	Amino Acid Change	RegDB Score‡	MA, MAF	Genotype	Genotype Count	Adjusted Mean (mg/dL)	SD (mg/dL)	β	SE	*P*	FDR§
SNPs with MAF ≥5%														
p1257	rs4238001	125348263	Exon 1	Gly2Ser	4	A, 0.0818	AA/GA/GG	5/91/521	123.77/137.71/137.08	25.91/48.56/39.43	-0.7889	4.1170	0.8481	0.997
p4072	rs7139401	125345448	Intron 1		3b	T, 0.4564	TT/CT/CC	136/293/190	139.69/135.86/136.87	45.96/39.26/37.36	1.2267	2.1986	0.5771	0.997
p5055	rs11057869	125344465	Intron 1		7	A, 0.0875	GG/GA/AA	514/98/5	137.41/135.96/123.77	39.57/47.36/25.91	-2.4528	4.0177	0.5418	0.997
p7650	rs11615630	125341870	Intron 1		5	A, 0.3817	GG/GA/AA	242/279/96	134.95/137.63/141.44	39.24/40.74/38.74	3.1377	2.2346	0.1608	0.997
p10292	rs4765181	125339228	Intron 1		5	T, 0.3972	TT/GT/GG	94/303/221	135.64/137.49/137.34	42.83/38.78/42.10	-0.6380	2.3317	0.7845	0.997
p16565	rs10773111	125332955	Intron 1		6	A, 0.4249	GG/GA/AA	203/306/110	139.77/136.67/134.18	39.41/40.38/43.14	-2.8406	2.2901	0.2153	0.997
p20207	rs11057853	125329313	Intron 1		5	G, 0.4269	GG/AG/AA	109/308/199	137.55/139.08/133.78	37.83/41.29/40.65	2.4118	2.3023	0.2953	0.997
p20741	rs11057851	125328779	Intron 1		5	T, 0.1157	TT/CT/CC	9/125/484	143.46/135.05/137.62	40.69/40.71/40.48	-1.2196	3.5190	0.7290	0.997
p21145	rs3924313	125328375	Intron 1		6	T, 0.3083	TT/CT/CC	52/277/289	141.41/139.43/134.64	31.85/40.42/41.13	3.9825	2.4728	0.1078	0.997
p22116	rs12370382	125327404	Intron 1		1f	A, 0.4005	GG/GA/AA	216/309/93	139.79/135.63/135.31	39.78/40.80/43.76	-2.6507	2.3677	0.2634	0.997
p26525	rs10773109	125322995	Intron 1		4	G, 0.4176	GG/CG/CC	102/313/204	142.24/135.46/137.13	45.30/39.07/41.37	1.8031	2.3509	0.4434	0.997
p28137	rs12229555	125321383	Intron 1		7	G, 0.2188	GG/AG/AA	31/208/378	143.82/135.28/137.45	47.88/41.04/40.40	0.2612	2.7448	0.9242	0.997
p28692	rs4765622	125320828	Intron 1		5	T, 0.4693	TT/CT/CC	136/308/174	137.29/134.95/140.45	40.81/40.26/42.18	-1.8314	2.2899	0.4241	0.997
p28957	rs11057844	125320563	Intron 1		5	A, 0.1839	GG/GA/AA	415/182/23	136.39/138.92/138.81	39.35/43.82/35.03	2.0109	2.8758	0.4846	0.997
p31072	rs10846749	125318448	Intron 1		4	G, 0.3448	CC/CG/GG	266/270/76	136.4/137.38/138.45	40.75/40.02/39.76	1.0276	2.3572	0.6630	0.997
p31938	rs10744182	125317582	Intron 1		5	G, 0.4130	GG/AC/AA	114/280/221	137.87/138.15/136.41	41.07/37.50/43.05	0.8915	2.2154	0.6875	0.997
p32129	rs10773107	125317391	Intron 1		7	T, 0.4828	TT/GT/GG	133/325/154	136.45/136.98/138.08	41.34/40.22/39.53	-0.8334	2.3295	0.7206	0.997
p32273	rs12580803	125317247	Intron 1		5	C, 0.1726	TT/TC/CC	420/181/16	137.04/137.05/137.91	42.80/36.98/26.72	0.1430	3.0524	0.9627	0.997
p32395	rs12581963	125317125	Intron 1		5	T, 0.0829	TT/CT/CC	5/93/523	163.79/135.45/137.28	77.60/39.28/40.32	1.0504	4.0479	0.7953	0.997
p32860	rs7967406	125316660	Intron 1		6	C, 0.3921	CC/AC/AA	98/291/232	140.78/134.77/138.47	36.62/40.80/42.23	0.1418	2.3043	0.9509	0.997
p36908	rs10846745	125312612	Intron 1		4	G, 0.4748	CC/CG/GG	171/305/140	139.13/134.92/139.3	40.88/40.72/40.81	-0.1277	2.2751	0.9553	0.997
p37095	rs10846744	125312425	Intron 1		4	G, 0.1364	CC/CG/GG	460/144/12	137.75/131.78/163.54	39.28/44.37/47.85	-0.6209	3.3079	0.8512	0.997
p42467	rs11057830	125307053	Intron 1		7	T, 0.1329	TT/CT/CC	12/140/465	152.41/135.21/137.51	56.28/41.90/39.45	0.5145	3.2852	0.8756	0.997
p48969	rs2343394	125300551	Intron 2		5	T, 0.2850	TT/CT/CC	55/240/319	137.58/137.48/136.42	35.95/41.26/40.84	0.7714	2.4591	0.7539	0.997
p49690	rs4765615	125299830	Intron 2		5	G, 0.4497	GG/AG/AA	120/314/182	133.24/139.08/135.79	42.48/40.69/40.27	-0.7893	2.3291	0.7348	0.997
p50151	rs2278986	125299369	Intron 3		5	C, 0.2890	CC/TC/TT	56/244/316	136.41/137.43/136.97	36.86/42.21/40.25	0.0131	2.4581	0.9957	0.997

p52556	rs11057820	125296964	Intron 4		5	G, 0.4871	GG/AG/AA	156/291/172	136.83/136.64/137.8	42.52/39.19/40.07	-0.5049	2.1745	0.8165	0.997
p55963	rs7134858	125293557	Intron 6		6	T, 0.0759	TT/CT/CC	7/80/532	143.07/143.43/135.6	41.19/34.29/41.32	6.7108	4.0972	0.1020	0.997
p60255	rs3782287	125289265	Intron 7		5	T, 0.4766	CC/CT/TT	176/296/147	139.48/134.45/140.24	34.27/43.14/41.85	0.1466	2.2118	0.9472	0.997
p63483	rs838912	125286037	Intron 7		7	A, 0.4927	AA/GA/GG	156/293/165	139.17/135.39/138.2	32.87/43.12/42.38	0.4413	2.2124	0.8420	0.997
p64772	rs5888	125284748	Exon 8	Ala350Ala	3a	T, 0.4846	CC/CT/TT	169/296/150	139.25/134.75/139.55	41.85/43.54/32.99	0.0119	2.2394	0.9958	0.997
p72551	rs1031605	125276969	Intron 9		5	T, 0.1755	CC/CT/TT	421/182/18	137.08/136.99/142.86	39.60/43.22/33.97	0.9119	2.9750	0.7593	0.997
p79721	rs838896	125269799	Intron 11		5	C, 0.3169	GG/GC/CC	293/257/67	137.07/134.13/148.32	40.37/42.24/35.30	2.7275	2.4011	0.2564	0.997
p79828	rs838895	125269692	Intron 11		5	G, 0.2891	GG/CG/CC	54/243/310	148.65/136.49/136.64	38.24/40.37/40.26	3.5525	2.4583	0.1489	0.997
p80045	rs838893	125269475	Intron 11		5	A, 0.2879	GG/GA/AA	316/251/53	136.65/134.96/147.53	40.04/41.98/35.23	2.4968	2.4732	0.3131	0.997
p83088	rs7977729	125266432	Intron 12		6	G, 0.2669	GG/AG/AA	44/243/333	145.57/137.07/135.99	32.22/42.84/40.42	3.0674	2.5689	0.2329	0.997
p83884	rs701106	125265636	Intron 12		5	T, 0.1527	TT/CT/CC	17/156/449	133.53/135.91/137.22	37.12/42.97/39.84	-1.4985	3.0783	0.6266	0.997
p87681	rs838883	125261839	3' flanking		5	A, 0.0845	GG/GA/AA	518/101/2	136.93/135.92/148.64	41.90/33.21/39.03	-0.4540	4.1635	0.9132	0.997
p87927	rs838880	125261593	3' flanking		5	G, 0.3237	AA/AG/GG	281/278/62	138.17/136.79/133.61	40.62/41.87/36.71	-1.9523	2.4487	0.4256	0.997
SNPs with MAF between 1-5%														
p13570	rs11057864	125335950	Intron 1		4	T, 0.0427	GT/GG	53/568	135.41/137.39	33.09/41.14	-1.9860	5.6944	0.7274	0.997
p49518	rs144194221	125300002	Intron 2		5	A, 0.0217	GA/GG	27/595	136.88/136.98	35.50/41.08	-0.0995	7.9121	0.9900	0.997
p54492	rs61762481	125295028	Intron 5		4	A, 0.0468	AA/GA/GG	3/52/564	134.57/143.36/136.36	17.20/41.80/40.76	5.4765	5.2143	0.2940	0.997
p56845	rs838902	125292675	Intron 6		5	G, 0.0211	AA/AG	589/26	137.03/130.98	40.32/50.18	-6.0568	7.9991	0.4492	0.997
p57508	rs71458866	125292012	Intron 7		4	A, 0.0162	GA/GG	20/596	141.97/136.96	46.68/40.62	5.0155	9.0820	0.5810	0.997
p57592	rs838903	125291928	Intron 7		4	A, 0.0161	GA/GG	20/600	142/136.99	46.68/40.63	5.0154	9.0821	0.5810	0.997
p58514	rs838905	125291006	Intron 7		4	C, 0.0347	TC/TT	43/577	132.91/137.47	45.10/40.48	-4.5621	6.3103	0.4700	0.997
p76757	rs9919713	125272763	Intron 9		6	T, 0.0379	AA/AT/TT	575/43/2	137.18/132.14/158.6	40.46/41.92/26.30	-2.4875	5.7658	0.6663	0.997
p77250	rs201901986	125272270	Intron 9		6	T, 0.0225	GG/GT	594/28	137.31/130.4	40.51/47.49	-6.9460	7.7359	0.3696	0.997
p77251	rs34339961	125272269	Intron 9		6	T, 0.0226	AA/AT	592/28	137.43/130.42	40.49/47.49	-7.0407	7.7372	0.3632	0.997
p77842	rs2272310	125271678	Intron 10		5	A, 0.0146	AA/GA/GG	1/16/599	166.93/137.23/136.81	NA/28.86/40.79	3.3863	8.9843	0.7064	0.997
p78430	rs838897	125271090	Intron 10		5	G, 0.0364	GG/CG/CC	2/41/575	158.56/132.37/137	26.30/42.64/40.52	-2.0316	5.8794	0.7298	0.997
p78747	rs2293440	125270773	Intron 11		5	C, 0.0243	CC/TC/TT	1/28/589	167.36/128.83/137.27	NA/48.00/40.43	-5.4568	7.2300	0.4507	0.997
SNPs with MAF ≤1%														
p1588		125347932	Intron 1		4	C, 0.0008	GC/GG	1/613	103.02/136.97	NA/40.77	-34.2394	40.0551	0.3930	0.997
p47235	rs199779577	125302285	Intron 1		4	T, 0.0016	CC/CT	615/2	137.09/129.72	40.84/0.28	-7.4285	28.3699	0.7935	0.997
p47416	rs73227571	125302104	Exon 2	Tyr92Tyr	5	T, 0.0008	CC/CT	622/1	137.14/43.3	40.68/NA	-94.8697	39.975	**0.0179**	0.619

p49978	rs5891	125299542	Exon 3	Val135Ile	5	A, 0.0096	GA/GG	12/611	123.95/137.25	41.99/40.78	-13.3163	11.6273	0.2525	0.997
p50432		125299088	Intron 3		2b	T, 0.0040	CC/CT	616/5	137.1/137.17	40.81/41.85	0.0726	17.9487	0.9968	0.997
p53093	rs201977189	125296427	Exon 5	Gly239Arg	5	A, 0.0016	GA/GG	2/615	162.96/136.79	31.11/40.83	26.3552	28.3632	0.3531	0.997
p53248	rs140613481	125296272	Intron 5		5	A, 0.0008	TA/TT	1/613	121.31/136.89	NA/40.65	-15.6212	39.821	0.6950	0.997
p54866		125294654	Intron 6		4	A, 0.0008	GA/GG	1/618	111.6/136.98	NA/40.78	-25.4825	39.9794	0.5241	0.997
p57308		125292212	Intron 7		4	T, 0.0016	CC/CT	615/2	137.16/107.71	40.77/34.37	-29.5184	28.2757	0.2969	0.997
p57618		125291902	Intron 7		4	A, 0.0008	GA/GG	1/617	164.16/136.85	NA/40.81	27.4039	39.9919	0.4935	0.997
p64285		125285235	Intron 7		3a	G, 0.0008	CC/CG	615/1	137.39/4.11	40.38/NA	-133.6798	39.6085	**0.0008**	0.054
p69612	rs147238482	125279908	Intron 8		4	A, 0.0048	GA/GG	6/617	129.04/137.07	19.82/40.96	-8.0419	16.3798	0.6236	0.997
p78334		125271186	Intron 10		4	G, 0.0024	TG/TT	3/620	164.85/136.86	22.11/40.84	28.0847	23.1060	0.2247	0.997
p86436	rs10396214	125263084	Exon 13-3' UTR	Arg484Trp[#]	4	T, 0.0088	CC/CT	612/11	136.98/137.5	40.39/62.06	0.5238	12.1954	0.9658	0.997
p87004	rs184715678	125262516	Exon 13-3' UTR		2b	A, 0.0016	CA/CC	2/619	154.66/137.04	55.44/40.78	17.6266	28.2688	0.5332	0.997
p87210		125262310	Exon 13-3' UTR		2b	A, 0.0016	GG/GA	621/2	137.04/122.1	40.86/8.49	-15.0364	28.3438	0.5960	0.997
p87381	rs192190977	125262139	3' flanking		2b	T, 0.0024	CC/CT	618/3	137.07/143.97	40.84/32.20	6.9443	23.1737	0.7645	0.997

FDR, false discovery rate; MA, minor allele; MAF, minor allele frequency; NA, not analyzed; RegDB, RegulomeDB; SD, standard deviation; SE, standard error; SNP, single nucleotide polymorphism; UTR, untranslated region.
All alleles on reverse strand. Results were adjusted for covariates: age and body mass index.
Significant *P*-values (*P* <0.05) are shown in **bold.**
*Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
†dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).
‡Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table A7 or can be seen at http://regulome.stanford.edu/help.
§FDR is referred to *q*-value in Benjamini-Hochberg procedure.
#rs10396214 is translated leading to amino acid change (Arg484Trp) in isoform 2 (alternative splice form); not translated in isoform 1.

Table A11. Single-site association results of 69 *SCARB1* genotyped variants for triglycerides (TG).

SNP Name*	SNP ID†	Chr12 Position*	Location	Amino Acid Change	RegDB Score‡	MA, MAF	Genotype	Genotype Count	Adjusted Mean (mg/dL)	SD (mg/dL)	β	SE	*P*	FDR§
SNPs with MAF ≥5%														
p1257	rs4238001	125348263	Exon 1	Gly2Ser	4	A, 0.0818	AA/GA/GG	5/90/518	136.99/134.49/138.6	42.45/66.44/66.04	-0.0201	0.0417	0.6295	0.991
p4072	rs7139401	125345448	Intron 1		3b	T, 0.4564	TT/CT/CC	135/291/190	138.31/135.33/140.8	65.54/66.92/62.90	-0.0140	0.0226	0.5353	0.991
p5055	rs11057869	125344465	Intron 1		7	A, 0.0875	GG/GA/AA	511/97/5	138.6/134.78/136.99	66.26/65.27/42.45	-0.0162	0.0407	0.6914	0.991
p7650	rs11615630	125341870	Intron 1		5	A, 0.3817	GG/GA/AA	241/278/96	139.18/136.16/138.93	64.71/67.33/64.50	-0.0046	0.0232	0.8428	0.991
p10292	rs4765181	125339228	Intron 1		5	T, 0.3972	TT/GT/GG	94/301/220	142.95/138.37/134.36	68.55/63.95/65.76	0.0312	0.0237	0.1884	0.896
p16565	rs10773111	125332955	Intron 1		6	A, 0.4249	GG/GA/AA	203/303/110	136.22/138.1/140.8	61.41/67.99/67.79	0.0087	0.0233	0.7098	0.991
p20207	rs11057853	125329313	Intron 1		5	G, 0.4269	GG/AG/AA	109/306/198	137.93/137.95/138.74	64.33/66.43/66.27	-0.0016	0.0235	0.9468	0.991
p20741	rs11057851	125328779	Intron 1		5	T, 0.1157	TT/CT/CC	9/124/482	125.25/131.35/139.79	46.08/63.07/66.57	-0.0536	0.0359	0.1357	0.851
p21145	rs3924313	125328375	Intron 1		6	T, 0.3083	TT/CT/CC	52/277/287	142.52/139.45/135.25	74.31/64.17/65.75	0.0249	0.0255	0.3297	0.991
p22116	rs12370382	125327404	Intron 1		1f	A, 0.4005	GG/GA/AA	216/305/93	137.38/138.65/136.36	63.78/67.64/63.72	-0.0060	0.0238	0.8024	0.991
p26525	rs10773109	125322995	Intron 1		4	G, 0.4176	GG/CG/CC	102/310/203	135.42/139.86/136.75	72.63/63.20/65.82	-0.0027	0.0236	0.9078	0.991
p28137	rs12229555	125321383	Intron 1		7	G, 0.2188	GG/AG/AA	31/207/375	141.64/134.74/139.28	73.37/60.72/67.22	-0.0086	0.0276	0.7556	0.991
p28692	rs4765622	125320828	Intron 1		5	T, 0.4693	TT/CT/CC	136/304/174	137.25/137.42/139	65.76/62.41/70.01	-0.0014	0.0230	0.9512	0.991
p28957	rs11057844	125320563	Intron 1		5	A, 0.1839	GG/GA/AA	414/180/23	137.59/138.48/137.95	64.47/66.24/85.51	-0.0071	0.0293	0.8088	0.991
p31072	rs10846749	125318448	Intron 1		4	G, 0.3448	CC/CG/GG	264/269/76	138.26/136.74/142.34	67.25/65.36/64.40	0.0114	0.0242	0.6390	0.991
p31938	rs10744182	125317582	Intron 1		5	G, 0.4130	GG/AC/AA	114/279/220	142.18/137.95/134.99	67.80/66.16/62.63	0.0182	0.0227	0.4238	0.991
p32129	rs10773107	125317391	Intron 1		7	T, 0.4828	TT/GT/GG	132/323/154	130.63/138.12/144.75	57.32/67.58/68.90	-0.0439	0.0239	0.0665	0.851
p32273	rs12580803	125317247	Intron 1		5	C, 0.1726	TT/TC/CC	417/180/16	135.67/141.36/160.43	65.45/66.32/69.97	0.0539	0.0308	0.0807	0.851
p32395	rs12581963	125317125	Intron 1		5	T, 0.0829	TT/CT/CC	5/92/521	182.53/133.59/138.11	85.61/60.00/66.52	0.0137	0.0413	0.7398	0.991
p32860	rs7967406	125316660	Intron 1		6	C, 0.3921	CC/AC/AA	98/288/231	128.28/139.7/140.48	60.92/67.37/65.59	-0.0322	0.0233	0.1668	0.886
p36908	rs10846745	125312612	Intron 1		4	G, 0.4748	CC/CG/GG	169/303/140	131.87/140.79/139.6	61.30/68.12/66.46	0.0206	0.0230	0.3706	0.991
p37095	rs10846744	125312425	Intron 1		4	G, 0.1364	CC/CG/GG	460/140/12	138.1/137.89/144.64	66.82/63.59/65.19	0.0170	0.0336	0.6132	0.991
p42467	rs11057830	125307053	Intron 1		7	T, 0.1329	TT/CT/CC	12/138/464	137.69/139.24/136.98	67.51/64.43/66.05	0.0207	0.0337	0.5394	0.991
p48969	rs2343394	125300551	Intron 2		5	T, 0.2850	TT/CT/CC	55/238/317	131.86/139.99/137.81	59.04/66.60/66.82	0.0031	0.0251	0.9015	0.991
p49690	rs4765615	125299830	Intron 2		5	G, 0.4497	GG/AG/AA	118/313/181	134.62/139.06/138.52	62.92/65.82/68.67	0.0007	0.0237	0.9779	0.991
p50151	rs2278986	125299369	Intron 3		5	C, 0.2890	CC/TC/TT	56/242/314	130.02/141.63/137.29	59.82/68.66/64.97	0.0020	0.0251	0.9377	0.991
p52556	rs11057820	125296964	Intron 4		5	G, 0.4871	GG/AG/AA	154/290/172	134.41/140.01/136.65	65.00/66.27/64.27	-0.0031	0.0224	0.8886	0.991

p55963	rs7134858	125293557	Intron 6		6	T, 0.0759	TT/CT/CC	7/80/528	151.11/133.61/138.08	74.24/63.08/66.12	-0.0070	0.0417	0.8663	0.991
p60255	rs3782287	125289265	Intron 7		5	T, 0.4766	CC/CT/TT	176/294/146	139.76/136.32/137.42	67.76/63.38/65.92	-0.0106	0.0224	0.6370	0.991
p63483	rs838912	125286037	Intron 7		7	A, 0.4927	AA/GA/GG	156/291/164	139.58/135.85/140.18	68.81/61.27/69.17	0.0003	0.0225	0.9905	0.991
p64772	rs5888	125284748	Exon 8	Ala350Ala	3a	T, 0.4846	CC/CT/TT	168/293/150	139.54/136.19/138.89	68.39/61.84/68.58	-0.0010	0.0225	0.9637	0.991
p72551	rs1031605	125276969	Intron 9		5	T, 0.1755	CC/CT/TT	419/181/18	138.85/138.59/105.34	63.59/71.47/44.68	-0.0518	0.0304	0.0885	0.851
p79721	rs838896	125269799	Intron 11		5	C, 0.3169	GG/GC/CC	292/254/67	135.97/140.69/136.52	65.03/68.61/58.96	0.0201	0.0243	0.4080	0.991
p79828	rs838895	125269692	Intron 11		5	G, 0.2891	GG/CG/CC	54/242/309	131.91/140.59/136.83	58.46/68.60/64.51	0.0060	0.0253	0.8131	0.991
p80045	rs838893	125269475	Intron 11		5	A, 0.2879	GG/GA/AA	315/248/53	136.86/140.45/135.57	63.98/68.48/64.09	0.0091	0.0252	0.7174	0.991
p83088	rs7977729	125266432	Intron 12		6	G, 0.2669	GG/AG/AA	44/240/332	138.3/138.25/138.21	66.10/65.82/65.89	0.0042	0.0261	0.8720	0.991
p83884	rs701106	125265636	Intron 12		5	T, 0.1527	TT/CT/CC	17/155/446	129.47/134.03/139.64	57.65/68.86/65.09	-0.0407	0.0313	0.1948	0.896
p87681	rs838883	125261839	3' flanking		5	A, 0.0845	GG/GA/AA	514/101/2	138.59/133.64/148.35	65.73/65.64/112.43	-0.0403	0.0422	0.3400	0.991
p87927	rs838880	125261593	3' flanking		5	G, 0.3237	AA/AG/GG	279/276/62	139.83/137.72/132.81	65.77/66.44/62.69	-0.0197	0.0247	0.4247	0.991
SNPs with MAF between 1-5%														
p13570	rs11057864	125335950	Intron 1		4	T, 0.0427	GT/GG	53/565	132.32/138.31	62.69/66.05	-0.0300	0.0579	0.6046	0.991
p49518	rs144194221	125300002	Intron 2		5	A, 0.0217	GA/GG	27/591	134.5/137.72	50.03/65.86	-0.0009	0.0796	0.9913	0.991
p54492	rs61762481	125295028	Intron 5		4	A, 0.0468	AA/GA/GG	3/52/560	187.79/138.77/137.71	77.57/64.22/66.05	0.0429	0.0528	0.4171	0.991
p56845	rs838902	125292675	Intron 6		5	G, 0.0211	AA/AG	585/26	136.75/158.22	64.55/83.90	0.1231	0.0805	0.1271	0.851
p57508	rs71458866	125292012	Intron 7		4	A, 0.0162	GA/GG	20/592	158.6/137.41	70.65/65.65	0.1461	0.0914	0.1102	0.851
p57592	rs838903	125291928	Intron 7		4	A, 0.0161	GA/GG	20/596	158.46/137.62	70.65/65.49	0.1431	0.0913	0.1175	0.851
p58514	rs838905	125291006	Intron 7		4	C, 0.0347	TC/TT	43/573	145.08/137.79	73.14/65.14	0.0514	0.0636	0.4190	0.991
p76757	rs9919713	125272763	Intron 9		6	T, 0.0379	AA/AT/TT	571/43/2	137.54/148.51/105.44	65.14/74.30/55.86	0.0439	0.0585	0.4535	0.991
p77250	rs201901986	125272270	Intron 9		6	T, 0.0225	GG/GT	590/28	137.12/152.11	64.82/81.72	0.0796	0.0779	0.3076	0.991
p77251	rs34339961	125272269	Intron 9		6	T, 0.0226	AA/AT	588/28	137.44/152.16	64.85/81.72	0.0774	0.0779	0.3210	0.991
p77842	rs2272310	125271678	Intron 10		5	A, 0.0146	AA/GA/GG	1/16/595	120.77/137.09/138.06	NA/58.90/66.19	0.0100	0.0913	0.9132	0.991
p78430	rs838897	125271090	Intron 10		5	G, 0.0364	GG/CG/CC	2/41/571	105.51/147.66/137.51	55.86/72.52/65.53	0.0439	0.0597	0.4626	0.991
p78747	rs2293440	125270773	Intron 11		5	C, 0.0243	CC/TC/TT	1/28/585	120.7/147.43/137.62	NA/79.99/65.30	0.0415	0.0732	0.5709	0.991
SNPs with MAF ≤1%														
p1588		125347932	Intron 1		4	C, 0.0008	GC/GG	1/609	114.33/138.04	NA/65.93	-0.1974	0.4051	0.6262	0.991
p47235	rs199779577	125302285	Intron 1		4	T, 0.0016	CC/CT	611/2	137.94/151.3	65.90/76.37	0.1488	0.2863	0.6034	0.991
p47416	rs73227571	125302104	Exon 2	Tyr92Tyr	5	T, 0.0008	CC/CT	618/1	138.04/70	65.75/NA	-0.5814	0.4054	0.1521	0.874
p49978	rs5891	125299542	Exon 3	Val135Ile	5	A, 0.0096	GA/GG	12/607	109.21/138.5	54.65/65.91	-0.2536	0.1170	**0.0306**	0.704

p50432		125299088	Intron 3		2b	T, 0.0040	CC/CT	612/5	138.3/123.05	65.88/45.14	-0.0369	0.1811	0.8387	0.991
p53093	rs201977189	125296427	Exon 5	Gly239Arg	5	A, 0.0016	GA/GG	2/611	105.79/138.31	10.61/65.99	-0.1647	0.2876	0.5671	0.991
p53248	rs140613481	125296272	Intron 5		5	A, 0.0008	TA/TT	1/609	264.51/137.61	NA/65.72	0.6474	0.4037	0.1093	0.851
p54866		125294654	Intron 6		4	A, 0.0008	GA/GG	1/614	106.57/138.1	NA/65.90	-0.2476	0.4056	0.5419	0.991
p57308		125292212	Intron 7		4	T, 0.0016	CC/CT	611/2	138.22/65.73	65.81/5.66	-0.6937	0.2844	**0.0150**	0.517
p57618		125291902	Intron 7		4	A, 0.0008	GA/GG	1/613	188.21/137.98	NA/66.00	0.4114	0.4055	0.3107	0.991
p64285		125285235	Intron 7		3a	G, 0.0008	CC/CG	611/1	137.69/406.12	64.94/NA	1.0938	0.4002	**0.0065**	0.446
p69612	rs147238482	125279908	Intron 8		4	A, 0.0048	GA/GG	6/613	128.85/138.02	64.56/65.82	-0.0667	0.1656	0.6870	0.991
p78334		125271186	Intron 10		4	G, 0.0024	TG/TT	3/616	163.77/137.81	73.08/65.78	0.2019	0.2337	0.3879	0.991
p86436	rs10396214	125263084	Exon 13-3' UTR	Arg484Trp[#]	4	T, 0.0088	CC/CT	609/10	138.14/125.27	65.99/46.94	-0.0630	0.1289	0.6251	0.991
p87004	rs184715678	125262516	Exon 13-3' UTR		2b	A, 0.0016	CA/CC	2/615	165.38/138.09	19.09/65.80	0.2703	0.2849	0.3432	0.991
p87210		125262310	Exon 13-3' UTR		2b	A, 0.0016	GG/GA	617/2	137.84/166.76	65.56/144.96	0.0583	0.2864	0.8389	0.991
p87381	rs192190977	125262139	3' flanking		2b	T, 0.0024	CC/CT	614/3	138.38/96.54	65.79/29.67	-0.2773	0.2334	0.2352	0.991

FDR, false discovery rate; MA, minor allele; MAF, minor allele frequency; NA, not analyzed; RegDB, RegulomeDB; SD, standard deviation; SE, standard error; SNP, single nucleotide polymorphism; UTR, untranslated region.
All alleles on reverse strand. TG values were Box-Cox transformed. Results were adjusted for covariates: sex, age, smoking and body mass index.
Significant *P*-values (*P* <0.05) are shown in **bold**.
*Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
†dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).
‡Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table A7 or can be seen at http://regulome.stanford.edu/help.
§FDR is referred to *q*-value in Benjamini-Hochberg procedure.
#rs10396214 is translated leading to amino acid change (Arg484Trp) in isoform 2 (alternative splice form); not translated in isoform 1.

Table A12. Significantly associated haplotype windows (global *P* <0.05) of 69 *SCARB1* genotyped variants with HDL-C, ApoB, LDL-C, and TG.

wind #	SNP 1-SNP 4 (SNP Name*-SNP ID†/ Chr12 Position*)	Chr12 Position*	Location	Amino Acid Change	Major/ Minor Alleles	MAF	β	Single site *P*	hap #	Haplotype	Hap Freq	Coef	SE	t stat	Hap *P*	Global *P*
HDL-C																
12	p22116-rs1230382	125327404	Intron 1		G/A	0.4005	-0.0031	0.7775	**h12.1**	GGGC	0.2078	0.0101	0.0144	0.7028	0.4824	0.0178
12	p26525-rs10773109	125322995	Intron 1		C/G	0.4176	-0.0132	0.2286	**h12.2**	GGAT	0.0221	0.0184	0.0391	0.4694	0.6389	
12	p28137-rs12229555	125321383	Intron 1		A/G	0.2188	0.0080	0.5310	**h12.3**	GGAC	0.1825	-0.0274	0.0151	-1.8131	0.0703	
12	p28692-rs4765622	125320828	Intron 1		C/T	0.4693	0.0130	0.2255	**h12.4**	GCAT	0.0585	0.0282	0.0243	1.1584	0.2471	
									h12.5	GCAC	0.1219	0.0089	0.0177	0.5045	0.6141	
									h12.6 (rare)	****	0.0175	-0.1146	0.0429	-2.6729	0.0077	
									hap base12	ACAT	0.3897	NA	NA	NA	NA	
14	p28137-rs12229555	125321383	Intron 1		A/G	0.2188	0.0080	0.5310	**h14.1**	GCGG	0.2176	-0.0048	0.0135	-0.3564	0.7216	0.0181
14	p28692-rs4765622	125320828	Intron 1		C/T	0.4693	0.0130	0.2255	**h14.2**	ACGG	0.1228	-0.0017	0.0171	-0.0984	0.9217	
14	p28957-rs11057844	125320563	Intron 1		G/A	0.1839	-0.0395	**0.0035**	**h14.3**	ACAC	0.1860	-0.0379	0.0143	-2.6557	0.0081	
14	p31072-rs10846749	125318448	Intron 1		C/G	0.3448	0.0059	0.5976	**h14.4 (rare)**	****	0.0041	0.1635	0.0920	1.7775	0.0760	
									hap base14	ATGC	0.4695	NA	NA	NA	NA	
15	p28692-rs4765622	125320828	Intron 1		C/T	0.4693	0.0130	0.2255	**h15.1**	TGCG	0.1891	-0.0412	0.0186	-2.2170	0.0270	0.0068
15	p28957-rs11057844	125320563	Intron 1		G/A	0.1839	-0.0395	**0.0035**	**h15.2**	CGGG	0.1356	0.0030	0.0189	0.1609	0.8722	
15	p31072-rs10846749	125318448	Intron 1		C/G	0.3448	0.0059	0.5976	**h15.3**	CGGA	0.2047	-0.0363	0.0178	-2.0387	0.0419	
15	p31938-rs10744182	125317582	Intron 1		A/G	0.4130	-0.0088	0.4065	**h15.4**	CACG	0.0876	-0.0564	0.0219	-2.5745	0.0103	
									h15.5	CACA	0.0983	-0.0555	0.0229	-2.4221	0.0157	
									h15.6 (rare)	****	0.0042	0.1431	0.0916	1.5621	0.1188	
									hap base 15	TGCA	0.2805	NA	NA	NA	NA	
16	p28957-rs11057844	125320563	Intron 1		G/A	0.1839	-0.0395	**0.0035**	**h16.1**	GCGT	0.0601	-0.0218	0.0301	-0.7249	0.4688	0.0092
16	p31072-rs10846749	125318448	Intron 1		C/G	0.3448	0.0059	0.5976	**h16.2**	GCGG	0.1284	-0.0495	0.0267	-1.8552	0.0640	
16	p31938-rs10744182	125317582	Intron 1		A/G	0.4130	-0.0088	0.4065	**h16.3**	GCAT	0.1377	0.0105	0.0258	0.4060	0.6849	
16	p32129-rs10773107	125317391	Intron 1		G/T	0.4828	-0.0140	0.2065	**h16.4**	GGGT	0.0412	-0.0811	0.0420	-1.9314	0.0539	
									h16.5	GGGG	0.0971	0.0479	0.0265	1.8059	0.0714	
									h16.6	GGAT	0.1255	-0.0407	0.0244	-1.6659	0.0962	
									h16.7	GGAG	0.0803	-0.0298	0.0322	-0.9234	0.3561	
									h16.8	ACGT	0.0391	-0.0126	0.0362	-0.3485	0.7276	
									h16.9	ACGG	0.0481	-0.0870	0.0338	-2.5745	0.0103	
									h16.10	ACAT	0.0787	-0.0722	0.0277	-2.6097	0.0093	
									h16.11	ACAG	0.0188	0.0123	0.0571	0.2158	0.8292	
									hap base 16	GCAG	0.1450	NA	NA	NA	NA	
60	p79828-rs838895	125269692	Intron 11		C/G	0.2891	0.0169	0.1489	**h60.1**	GAGT	0.0115	0.1470	0.0671	2.1920	0.0288	0.0453
60	p80045-rs838893	125269475	Intron 11		G/A	0.2879	0.0136	0.2472	**h60.2**	GAGC	0.2497	0.0095	0.0129	0.7327	0.4640	
60	p83088-rs7977729	125266432	Intron 12		A/G	0.2669	0.0107	0.3768	**h60.3**	GAAT	0.0175	0.0557	0.0416	1.3393	0.1810	
60	p83884-rs701106	125265636	Intron 12		C/T	0.1527	0.0394	**0.0066**	**h60.4**	CGAT	0.1146	0.0329	0.0170	1.9359	0.0533	
									h60.5 (rare)	****	0.0244	0.0212	0.0338	0.6281	0.5302	
									hap base 60	CGAC	0.5822	NA	NA	NA	NA	
61	p80045-rs838893	125269475	Intron 11		G/A	0.2879	0.0136	0.2472	**h61.1**	GATC	0.1147	0.0366	0.0169	2.1674	0.0306	0.0236

61	p83088-rs7977729	125266432	Intron 12		A/G	0.2669	0.0107	0.3768	**h61.2**	AGTC	0.0118	0.1512	0.0644	2.3472	0.0192	
61	p83884-rs701106	125265636	Intron 12		C/T	0.1527	0.0394	**0.0066**	**h61.3**	AGCC	0.2520	0.0086	0.0128	0.6715	0.5021	
61	p86436-rs10396214	125263084	Exon 13-3' UTR	Arg484Trp[#]	C/T	0.0088	-0.0402	0.4808	**h61.4**	AATC	0.0170	0.0577	0.0432	1.3354	0.1822	
									h61.5 (rare)	****	0.0185	0.0221	0.0384	0.5752	0.5653	
									hap base 61	GACC	0.5860	NA	NA	NA	NA	
62	p83088-rs7977729	125266432	Intron 12		A/G	0.2669	0.0107	0.3768	**h62.1**	GTCC	0.0133	0.1782	0.0617	2.8882	0.0040	0.0034
62	p83884-rs701106	125265636	Intron 12		C/T	0.1527	0.0394	**0.0066**	**h62.2**	GCCC	0.2520	0.0086	0.0128	0.6714	0.5022	
62	p86436-rs10396214	125263084	Exon 13-3' UTR	Arg484Trp[#]	C/T	0.0088	-0.0402	0.4808	**h62.3**	ATCC	0.1302	0.0349	0.0162	2.1529	0.0317	
62	p87004-rs184715678	125262516	Exon 13-3' UTR		C/A	0.0016	-0.2166	0.1019	**h62.4 (rare)**	****	0.0105	-0.0572	0.0515	-1.1117	0.2667	
									hap base 62	ACCC	0.5940	NA	NA	NA	NA	
63	p83884-rs701106	125265636	Intron 12		C/T	0.1527	0.0394	**0.0066**	**h63.1**	TCCG	0.1436	0.0432	0.0147	2.9327	0.0035	0.0047
63	p86436-rs10396214	125263084	Exon 13-3' UTR	Arg484Trp[#]	C/T	0.0088	-0.0402	0.4808	**h63.2 (rare)**	****	0.0121	-0.0643	0.0482	-1.3328	0.1831	
63	p87004-rs184715678	125262516	Exon 13-3' UTR		C/A	0.0016	-0.2166	0.1019	**hap base 63**	CCCG	0.8444	NA	NA	NA	NA	
63	p87210-chr12_125262310	125262310	Exon 13-3' UTR		G/A	0.0016	-0.0914	0.4911								
ApoB																
21	p32395-rs12581963	125317125	Intron 1		C/T	0.0829	-0.8103	0.2986	**h21.1**	TACG	0.0836	-0.2165	0.8205	-0.2638	0.7920	0.0363
21	p32860-rs7967406	125316660	Intron 1		A/C	0.3921	0.7558	0.0920	**h21.2**	CCCC	0.4101	1.0295	0.4756	2.1647	0.0310	
21	p36908-rs10846745	125312612	Intron 1		C/G	0.4748	-0.8517	0.0551	**h21.3**	CACG	0.0489	2.5189	1.0809	2.3305	0.0202	
21	p37095-rs10846744	125312425	Intron 1		C/G	0.1364	0.0774	0.9027	**h21.4 (rare)**	****	0.0095	-1.9067	2.3286	-0.8188	0.4133	
									hap base 21	CAGC	0.4480	NA	NA	NA	NA	
25	p42467-rs11057830	125307053	Intron 1		C/T	0.1329	0.1439	0.8205	**h25.1**	TCCC	0.1359	0.5399	0.6521	0.8280	0.4081	0.0341
25	p47235-rs199779577	125302285	Intron 1		C/T	0.0016	2.1039	0.7471	**h25.2**	CCCT	0.2988	1.3478	0.4944	2.7259	0.0067	
25	p47416-rs73227571	125302104	Exon 2	Tyr92Tyr	C/T	0.0008	-12.3322	0.0599	**h25.3 (rare)**	****	0.0023	-4.1374	4.5850	-0.9024	0.3674	
25	p48969-rs2343394	125300551	Intron 2		C/T	0.2850	1.2544	**0.0082**	**hap base 25**	CCCC	0.5629	NA	NA	NA	NA	
26	p47235-rs199779577	125302285	Intron 1		C/T	0.0016	2.1039	0.7471	**h26.1**	CCTG	0.2983	1.2602	0.4792	2.6298	0.0089	0.0293
26	p47416-rs73227571	125302104	Exon 2	Tyr92Tyr	C/T	0.0008	-12.3322	0.0599	**h26.2**	CCCA	0.0231	1.5759	1.4749	1.0685	0.2859	
26	p48969-rs2343394	125300551	Intron 2		C/T	0.2850	1.2544	**0.0082**	**h26.3 (rare)**	****	0.0023	-4.2638	4.5761	-0.9318	0.3520	
26	p49518-rs144194221	125300002	Intron 2		G/A	0.0217	1.4134	0.3456	**hap base 26**	CCCG	0.6763	NA	NA	NA	NA	
27	p47416-rs73227571	125302104	Exon 2	Tyr92Tyr	C/T	0.0008	-12.3322	0.0599	**h27.1**	CTGG	0.2953	1.4591	0.5012	2.9112	0.0038	0.0465
27	p48969-rs2343394	125300551	Intron 2		C/T	0.2850	1.2544	**0.0082**	**h27.2**	CCAG	0.0218	1.9730	1.5342	1.2860	0.1991	
27	p49518-rs144194221	125300002	Intron 2		G/A	0.0217	1.4134	0.3456	**h27.3**	CCGG	0.1470	0.7838	0.6504	1.2050	0.2289	
27	p49690-rs4765615	125299830	Intron 2		A/G	0.4497	1.2493	**0.0059**	**h27.4 (rare)**	****	0.0040	1.7464	3.6393	0.4799	0.6316	
									hap base 27	CCGA	0.5319	NA	NA	NA	NA	
28	p48969-rs2343394	125300551	Intron 2		C/T	0.2850	1.2544	**0.0082**	**h28.1**	TGGG	0.2954	1.5188	0.4952	3.0670	0.0023	0.0005
28	p49518-rs144194221	125300002	Intron 2		G/A	0.0217	1.4134	0.3456	**h28.2**	CAGG	0.0219	2.1453	1.5141	1.4169	0.1572	
28	p49690-rs4765615	125299830	Intron 2		A/G	0.4497	1.2493	**0.0059**	**h28.3**	CGGG	0.1467	0.9193	0.6438	1.4279	0.1540	
28	p49978-rs5891	125299542	Exon 3	Val135Ile	G/A	0.0096	5.8266	**0.0120**	**h28.4 (rare)**	****	0.0120	6.6169	2.0468	3.2327	0.0013	
									hap base 28	CGAG	0.5240	NA	NA	NA	NA	
29	p49518-rs144194221	125300002	Intron 2		G/A	0.0217	1.4134	0.3456	**h29.1**	AGAC	0.0218	2.0500	1.5295	1.3403	0.1809	0.0031

29	p49690-rs4765615	125299830	Intron 2		A/G	0.4497	1.2493	**0.0059**	**h29.2**	GGGC	0.3003	1.4871	0.4933	3.0147	0.0027	
29	p49978-rs5891	125299542	Exon 3	Val135Ile	G/A	0.0096	5.8266	**0.0120**	**h29.3**	GGGT	0.1417	0.8811	0.6642	1.3265	0.1854	
29	p50151-rs2278986	125299369	Intron 3		T/C	0.2890	1.1926	**0.0122**	**h29.4 (rare)**	****	0.0121	5.2239	2.0469	2.5521	0.0111	
									hap base 29	GAGT	0.5241	NA	NA	NA	NA	
30	p49690-rs4765615	125299830	Intron 2		A/G	0.4497	1.2493	**0.0059**	**h30.1**	GGCC	0.2994	1.5090	0.4951	3.0481	0.0024	0.0021
30	p49978-rs5891	125299542	Exon 3	Val135Ile	G/A	0.0096	5.8266	**0.0120**	**h30.2**	GGTC	0.1638	1.0307	0.6363	1.6200	0.1060	
30	p50151-rs2278986	125299369	Intron 3		T/C	0.2890	1.1926	**0.0122**	**h30.3 (rare)**	****	0.0127	4.6601	1.9603	2.3772	0.0179	
30	p50432-chr12_125299088	125299088	Intron 3		C/T	0.0040	0.2876	0.9396	**hap base 30**	AGTC	0.5240	NA	NA	NA	NA	
31	p49978-rs5891	125299542	Exon 3	Val135Ile	G/A	0.0096	5.8266	**0.0120**	**h31.1**	GCCG	0.2970	1.3482	0.4989	2.7026	0.0072	0.0027
31	p50151-rs2278986	125299369	Intron 3		T/C	0.2890	1.1926	**0.0122**	**h31.2**	GTCG	0.2004	0.4892	0.5601	0.8733	0.3830	
31	p50432-chr12_125299088	125299088	Intron 3		C/T	0.0040	0.2876	0.9396	**h31.3 (rare)**	****	0.0156	4.9815	1.8194	2.7380	0.0064	
31	p52556-rs11057820	125296964	Intron 4		A/G	0.4871	0.8700	**0.0436**	**hap base 31**	GTCA	0.4870	NA	NA	NA	NA	
44	p57618-chr12_125291902	125291902	Intron 7		NA	NA	NA	NA	**h44.1**	[NA]CCA	0.0116	0.4765	2.0887	0.2281	0.8197	0.0459
44	p58514-rs838905	125291006	Intron 7		T/C	0.0347	-2.0859	0.0914	**h44.2**	[NA]CCG	0.0231	-3.4968	1.4928	-2.3424	0.0196	
44	p60255-rs3782287	125289265	Intron 7		C/T	0.4766	-0.2158	0.6229	**h44.3**	[NA]TTG	0.4722	-0.3250	0.4387	-0.7407	0.4593	
44	p63483-rs838912	125286037	Intron 7		G/A	0.4927	0.3773	0.3856	**h44.4 (rare)**	****	0.0084	4.5791	2.4591	1.8621	0.0633	
									hap base 44	[NA]TCA	0.4848	NA	NA	NA	NA	
53	p77250-rs201901986	125272270	Intron 9		G/T	0.0225	-1.1574	0.4385	**h53.1**	GAAT	0.0152	0.3683	1.6971	0.2170	0.8283	0.0075
53	p77251-rs34339961	125272269	Intron 9		A/T	0.0226	-1.1124	0.4559	**h53.2**	TTGT	0.0219	-1.9861	1.5059	-1.3189	0.1879	
53	p77842-rs2272310	125271678	Intron 10		G/A	0.0146	0.3875	0.8187	**h53.3 (rare)**	****	0.0023	14.4708	4.5507	3.1799	0.0016	
53	p78334-chr12_125271186	125271186	Intron 10		T/G	0.0024	14.5804	**0.0015**	**hap base 53**	GAGT	0.9606	NA	NA	NA	NA	
54	p77251-rs34339961	125272269	Intron 9		A/T	0.0226	-1.1124	0.4559	**h54.1**	AATG	0.0138	0.4249	1.7520	0.2426	0.8085	0.0487
54	p77842-rs2272310	125271678	Intron 10		G/A	0.0146	0.3875	0.8187	**h54.2**	TGTG	0.0208	-2.2378	1.5539	-1.4401	0.1506	
54	p78334-chr12_125271186	125271186	Intron 10		T/G	0.0024	14.5804	**0.0015**	**h54.3 (rare)**	****	0.0048	7.6445	3.2455	2.3554	0.0190	
54	p78430-rs838897	125271090	Intron 10		C/G	0.0364	-0.6962	0.5598	**hap base 54**	AGTC	0.9607	NA	NA	NA	NA	
LDL-C																
1	p1257-rs4238001	125348263	Exon 1	Gly2Ser	G/A	0.0818	-0.7889	0.8481	**h1.1**	AGTA	0.0802	0.0666	4.1962	0.0159	0.9873	0.0387
1	p1588-chr12_125347932	125347932	Intron 1		G/C	0.0008	-34.2394	0.3930	**h1.2**	GGTG	0.3741	1.5753	2.3906	0.6589	0.5102	
1	p4072-rs7139401	125345448	Intron 1		C/T	0.4564	1.2267	0.5771	**h1.3 (rare)**	****	0.0082	-35.2873	15.5864	-2.2640	0.0239	
1	p5055-rs11057869	125344465	Intron 1		G/A	0.0875	-2.4528	0.5418	**hap base 1**	GGCG	0.5375	NA	NA	NA	NA	
39	p55963-rs7134858	125293557	Intron 6		C/T	0.0759	6.7108	0.1020	**h39.1**	TACG	0.0759	6.2462	4.1005	1.5233	0.1282	0.0168
39	p56845-rs838902	125292675	Intron 6		A/G	0.0211	-6.0568	0.4492	**h39.2**	CGCA	0.0161	4.8552	8.9774	0.5408	0.5888	
39	p57308-chr12_125292212	125292212	Intron 7		C/T	0.0016	-29.5184	0.2969	**h39.3 (rare)**	****	0.0065	-38.2029	0.2334	-163.6845	<1.00E-06	
39	p57508-rs71458866	125292012	Intron 7		G/A	0.0162	5.0155	0.5810	**hap base 39**	CACG	0.9015	NA	NA	NA	NA	
40	p56845-rs838902	125292675	Intron 6		A/G	0.0211	-6.0568	0.4492	**h40.1**	GCAA	0.0161	4.5712	8.9925	0.5083	0.6114	0.0178
40	p57308-chr12_125292212	125292212	Intron 7		C/T	0.0016	-29.5184	0.2969	**h40.2 (rare)**	****	0.0065	-39.1903	0.2102	-186.4715	<1.00E-06	
40	p57508-rs71458866	125292012	Intron 7		G/A	0.0162	5.0155	0.5810	**hap base 40**	ACGG	0.9775	NA	NA	NA	NA	
40	p57592-rs838903	125291928	Intron 7		G/A	0.0161	5.0154	0.5810								
TG																
16	p28957-rs11057844	125320563	Intron 1		G/A	0.1839	-0.0071	0.8088	**h16.1**	GCGT	0.0573	-0.0718	0.0671	-1.0708	0.2847	0.0315

16	p31072-rs10846749	125318448	Intron 1	C/G	0.3448	0.0114	0.6390	**h16.2**	GCGG	0.1284	0.0880	0.0644	1.3652	0.1727	
16	p31938-rs10744182	125317582	Intron 1	A/G	0.4130	0.0182	0.4238	**h16.3**	GCAT	0.1401	-0.0027	0.0613	-0.0437	0.9652	
16	p32129-rs10773107	125317391	Intron 1	G/T	0.4828	-0.0439	0.0665	**h16.4**	GGGT	0.0432	0.1226	0.0858	1.4280	0.1538	
								h16.5	GGGG	0.0966	-0.0942	0.0579	-1.6267	0.1043	
								h16.6	GGAT	0.1248	0.0382	0.0521	0.7341	0.4632	
								h16.7	GGAG	0.0812	0.0782	0.0720	1.0860	0.2779	
								h16.8	ACGT	0.0400	-0.0402	0.0813	-0.4946	0.6211	
								h16.9	ACGG	0.0476	0.2008	0.0749	2.6793	0.0076	
								h16.10	ACAT	0.0766	-0.0815	0.0664	-1.2270	0.2203	
								h16.11	ACAG	0.0195	0.0494	0.1369	0.3606	0.7186	
								hap base 16	GCAG	0.1448	NA	NA	NA	NA	
17	p31072-rs10846749	125318448	Intron 1	C/G	0.3448	0.0114	0.6390	**h17.1**	CGTT	0.0874	0.0186	0.0708	0.2625	0.7930	0.0100
17	p31938-rs10744182	125317582	Intron 1	A/G	0.4130	0.0182	0.4238	**h17.2**	CGGT	0.1399	0.0987	0.0461	2.1413	0.0327	
17	p32129-rs10773107	125317391	Intron 1	G/T	0.4828	-0.0439	0.0665	**h17.3**	CGGC	0.0349	0.3546	0.0917	3.8692	0.0001	
17	p32273-rs12580803	125317247	Intron 1	T/C	0.1726	0.0539	0.0807	**h17.4**	CATC	0.0229	0.0565	0.1129	0.5007	0.6167	
								h17.5	CAGT	0.1168	0.0607	0.0677	0.8964	0.3704	
								h17.6	CAGC	0.0485	0.0727	0.0703	1.0330	0.3020	
								h17.7	GGTT	0.0431	0.1694	0.0826	2.0510	0.0407	
								h17.8	GGGT	0.0552	-0.0816	0.0743	-1.0978	0.2727	
								h17.9	GGGC	0.0413	0.0090	0.0711	0.1260	0.8997	
								h17.10	GATT	0.1222	0.0953	0.0568	1.6763	0.0942	
								h17.11	GAGT	0.0695	0.0878	0.0601	1.4604	0.1447	
								h17.12	GAGC	0.0112	0.2559	0.1547	1.6544	0.0986	
								h17.13 (rare)	****	0.0142	-0.1698	0.1341	-1.2664	0.2058	
								hap base 17	CATT	0.1930	NA	NA	NA	NA	

ApoB, apolipoprotein B; Coef, coefficient; HDL-C, high-density lipoprotein cholesterol; LDL-C, low-density lipoprotein cholesterol; MAF, minor allele frequency; NA, not analyzed; SE, standard error; SNP, single nucleotide polymorphism; TG, triglycerides; UTR, untranslated region.

All alleles on reverse strand. HDL-C, ApoB, and TG values were Box-Cox transformed. Results were adjusted for covariates: sex, age, smoking, and body mass index.

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.

For SNPs with MAF ≥5%, significant single-site *P*-values (*P* <0.05; Tables A8-A11) are shown in **bold**.

Detailed haplotype association results of all haplotype windows for each trait are shown in Tables A14 and A15; see haplotype association plots in Figure A5.

*Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.

†dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).

#rs10396214 is translated leading to amino acid change (Arg484Trp) in isoform 2 (alternative splice form); not translated in isoform 1.

Table A13. Summary of *SCARB1* consecutive haplotype windows and regions significantly associated with HDL-C (regions: 1, 4) and ApoB (regions: 2, 3; global *P* <0.05, see haplotype association plots and regions in Figure 6.2).

Region #	Trait	Consecutive Significantly Associated Haplotype Windows (global *P* <0.05)*							
		Haplotype Window	Chr12 Position† (Location)		The Composited Variants from 5' to 3' Direction			Most Relevant Haplotype	
		wind #	Start (5')	End (3')	SNP Name† - SNP ID‡/Chr12 Position†	Major/Minor Alleles	hap #	Sequence	β (range: min-max)
1	HDL-C	14	125327404	125317391	p28137-rs1229555	G/A	h14.3	ACACGG	(-0.0870)-(-0.0379)
		15	(intron 1)	(intron 1)	p28692-rs4765622	C/G	h15.4		
		16			**p28957-rs11057844**	A/G	h16.9		
					p31072-rs10846749	C/G			
					p31938-rs10744192	A/G			
					p32129-rs10773107	G/T			
2	ApoB	25	125307053	125296964	p42467-rs11057830	C/T	h25.2	CCCTGGGCCG	1.2602-1.5188
		26	(intron 1)	(intron 4)	p47235-rs199779577	C/T	h26.1		
		27			p47416-rs73227571(Tyr92Tyr)	C/T	h27.1		
		28			**p48969-rs2343394**	C/T	h28.1		
		29			p49518-rs144194221	G/A	h29.2		
		30			**p49690-rs4765615**	A/G	h30.1		
		31			p49978-rs5891(Val135Ile)	G/A	h31.1		
					p50151-rs2278986	T/C			
					p50432-chr12_125299088	C/T			
					p52556-rs11057820	A/G			
3	ApoB	53	125272270	125271090	p77250-rs201901986	G/A	h53.3	***** (rare; hap freq <1%)	7.6445-14.4708
		54	(intron 9)	(intron 10)	p77251-rs34339961	A/T	h54.3		
					p77842-rs2272310	G/A			
					p78334-chr12_125271186	T/G			
					p78430-rs838897	C/G			
4	HDL-C	60	125269692	125262310 (exon 13-	p79828-rs838895	C/G	h60.1	GAGTCCG	0.0432-0.1782
		61	(intron 11)	3' UTR)	p80045-rs838893	G/A	h61.2		
		62			p83088-rs797729	A/G	h62.1		
		63			**p83884-rs701106**	C/T	h63.1		
					p86436-rs10396214(Arg484Trp)§	C/T			
					p87004-rs184715678	C/A			
					p87210-chr12_125262310	G/A			

ApoB, apolipoprotein B; HDL-C, high-density lipoprotein cholesterol; SNP, single nucleotide polymorphism; UTR, untranslated region.
All alleles on reverse strand. HDL-C and ApoB values were Box-Cox transformed. Results were adjusted for covariates (HDL-C: sex, age, smoking, and body mass index; ApoB: age and smoking).
The composited variants in each region are shown as "SNP Name-SNP ID/Chr12 Position (for novel variants)", and corresponding to 5' to 3' direction.
Each variant is presented as "SNP Name-SNP ID/Chr12 Position (for novel variants)".
SNPs with MAF ≥5% that yielded a significance evidence of single-site association (*P* <0.05; Tables A8-A11) observed in this study are in **bold**.
*Detailed haplotype windows significantly associated with HDL-C and ApoB (global *P* <0.05) can be seen in Tables A12 and A14.
†Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
‡dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).
§rs10396214 is translated leading to Arg484Trp in isoform 2 (alternative splice form); not translated in isoform 1.

Table A14. Haplotype association results of 69 *SCARB1* genotyped variants (haplotype windows and global *P*-values) for HDL-C and ApoB.

						HDL-C				ApoB			
wind #	wind SNP	SNP Name*-SNP ID†/ Chr12 Position*	Location	Genotype	MA, MAF	GT Count	β	Single site *P*	Global *P*	GT Count	β	Single site *P*	Global *P*
1	SNP 1	p1257-rs4238001	Exon 1	AA/GA/GG	A, 0.0818	5/91/519	0.0224	0.2455	0.5885	5/64/360	0.2786	0.7161	0.5091
1	SNP 2	p1588-chr12_125347932	Intron 1	GC/GG	C, 0.0008	1/611	0.3089	0.1008		1/426	-8.6447	0.1806	
1	SNP 3	p4072-rs7139401	Intron 1	TT/CT/CC	T, 0.4564	135/293/189	0.0097	0.3552		91/199/142	0.4818	0.2657	
1	SNP 4	p5055-rs11057869	Intron 1	GG/GA/AA	A, 0.0875	512/98/5	0.0220	0.2433		353/71/5	0.1155	0.8766	
2	SNP 1	p1588-chr12_125347932	Intron 1	GC/GG	C, 0.0008	1/611	0.3089	0.1008	0.6095	1/426	-8.6447	0.1806	0.5792
2	SNP 2	p4072-rs7139401	Intron 1	TT/CT/CC	T, 0.4564	135/293/189	0.0097	0.3552		91/199/142	0.4818	0.2657	
2	SNP 3	p5055-rs11057869	Intron 1	GG/GA/AA	A, 0.0875	512/98/5	0.0220	0.2433		353/71/5	0.1155	0.8766	
2	SNP 4	p7650-rs11615630	Intron 1	GG/GA/AA	A, 0.3817	241/279/95	0.0006	0.9545		181/192/59	0.7301	0.1080	
3	SNP 1	p4072-rs7139401	Intron 1	TT/CT/CC	T, 0.4564	135/293/189	0.0097	0.3552	0.8713	91/199/142	0.4818	0.2657	0.3695
3	SNP 2	p5055-rs11057869	Intron 1	GG/GA/AA	A, 0.0875	512/98/5	0.0220	0.2433		353/71/5	0.1155	0.8766	
3	SNP 3	p7650-rs11615630	Intron 1	GG/GA/AA	A, 0.3817	241/279/95	0.0006	0.9545		181/192/59	0.7301	0.1080	
3	SNP 4	p10292-rs4765181	Intron 1	TT/GT/GG	T, 0.3972	93/303/220	-0.0074	0.5047		75/211/146	-0.0530	0.9069	
4	SNP 1	p5055-rs11057869	Intron 1	GG/GA/AA	A, 0.0875	512/98/5	0.0220	0.2433	0.8911	353/71/5	0.1155	0.8766	0.4445
4	SNP 2	p7650-rs11615630	Intron 1	GG/GA/AA	A, 0.3817	241/279/95	0.0006	0.9545		181/192/59	0.7301	0.1080	
4	SNP 3	p10292-rs4765181	Intron 1	TT/GT/GG	T, 0.3972	93/303/220	-0.0074	0.5047		75/211/146	-0.0530	0.9069	
4	SNP 4	p13570-rs11057864	Intron 1	GT/GG	T, 0.0427	53/566	0.0236	0.3795		43/390	0.5236	0.6186	
5	SNP 1	p7650-rs11615630	Intron 1	GG/GA/AA	A, 0.3817	241/279/95	0.0006	0.9545	0.9005	181/192/59	0.7301	0.1080	0.1824
5	SNP 2	p10292-rs4765181	Intron 1	TT/GT/GG	T, 0.3972	93/303/220	-0.0074	0.5047		75/211/146	-0.0530	0.9069	
5	SNP 3	p13570-rs11057864	Intron 1	GT/GG	T, 0.0427	53/566	0.0236	0.3795		43/390	0.5236	0.6186	
5	SNP 4	p16565-rs10773111	Intron 1	GG/GA/AA	A, 0.4249	202/306/109	-0.0011	0.9160		139/209/84	-0.6627	0.1356	
6	SNP 1	p10292-rs4765181	Intron 1	TT/GT/GG	T, 0.3972	93/303/220	-0.0074	0.5047	0.1858	75/211/146	-0.0530	0.9069	0.2227
6	SNP 2	p13570-rs11057864	Intron 1	GT/GG	T, 0.0427	53/566	0.0236	0.3795		43/390	0.5236	0.6186	
6	SNP 3	p16565-rs10773111	Intron 1	GG/GA/AA	A, 0.4249	202/306/109	-0.0011	0.9160		139/209/84	-0.6627	0.1356	
6	SNP 4	p20207-rs11057853	Intron 1	GG/AG/AA	G, 0.4269	109/307/198	-0.0047	0.6641		72/209/149	0.8207	0.0680	
7	SNP 1	p13570-rs11057864	Intron 1	GT/GG	T, 0.0427	53/566	0.0236	0.3795	0.1880	43/390	0.5236	0.6186	0.5036
7	SNP 2	p16565-rs10773111	Intron 1	GG/GA/AA	A, 0.4249	202/306/109	-0.0011	0.9160		139/209/84	-0.6627	0.1356	
7	SNP 3	p20207-rs11057853	Intron 1	GG/AG/AA	G, 0.4269	109/307/198	-0.0047	0.6641		72/209/149	0.8207	0.0680	
7	SNP 4	p20741-rs11057851	Intron 1	TT/CT/CC	T, 0.1157	9/125/482	0.0073	0.6597		7/86/339	0.7897	0.2529	
8	SNP 1	p16565-rs10773111	Intron 1	GG/GA/AA	A, 0.4249	202/306/109	-0.0011	0.9160	0.0685	139/209/84	-0.6627	0.1356	0.2269
8	SNP 2	p20207-rs11057853	Intron 1	GG/AG/AA	G, 0.4269	109/307/198	-0.0047	0.6641		72/209/149	0.8207	0.0680	
8	SNP 3	p20741-rs11057851	Intron 1	TT/CT/CC	T, 0.1157	9/125/482	0.0073	0.6597		7/86/339	0.7897	0.2529	
8	SNP 4	p21145-rs3924313	Intron 1	TT/CT/CC	T, 0.3083	52/275/289	-0.0101	0.3898		36/184/212	0.6565	0.1821	
9	SNP 1	p20207-rs11057853	Intron 1	GG/AG/AA	G, 0.4269	109/307/198	-0.0047	0.6641	0.4096	72/209/149	0.8207	0.0680	0.1234
9	SNP 2	p20741-rs11057851	Intron 1	TT/CT/CC	T, 0.1157	9/125/482	0.0073	0.6597		7/86/339	0.7897	0.2529	
9	SNP 3	p21145-rs3924313	Intron 1	TT/CT/CC	T, 0.3083	52/275/289	-0.0101	0.3898		36/184/212	0.6565	0.1821	
9	SNP 4	p22116-rs1230382	Intron 1	GG/GA/AA	A, 0.4005	215/308/93	-0.0031	0.7775		143/215/73	-0.0729	0.8730	
10	SNP 1	p20741-rs11057851	Intron 1	TT/CT/CC	T, 0.1157	9/125/482	0.0073	0.6597	0.2408	7/86/339	0.7897	0.2529	0.2684
10	SNP 2	p21145-rs3924313	Intron 1	TT/CT/CC	T, 0.3083	52/275/289	-0.0101	0.3898		36/184/212	0.6565	0.1821	
10	SNP 3	p22116-rs1230382	Intron 1	GG/GA/AA	A, 0.4005	215/308/93	-0.0031	0.7775		143/215/73	-0.0729	0.8730	
10	SNP 4	p26525-rs10773109	Intron 1	GG/CG/CC	G, 0.4176	102/312/203	-0.0132	0.2286		67/211/154	0.0192	0.9667	
11	SNP 1	p21145-rs3924313	Intron 1	TT/CT/CC	T, 0.3083	52/275/289	-0.0101	0.3898	0.1356	36/184/212	0.6565	0.1821	0.6796
11	SNP 2	p22116-rs1230382	Intron 1	GG/GA/AA	A, 0.4005	215/308/93	-0.0031	0.7775		143/215/73	-0.0729	0.8730	
11	SNP 3	p26525-rs10773109	Intron 1	GG/CG/CC	G, 0.4176	102/312/203	-0.0132	0.2286		67/211/154	0.0192	0.9667	
11	SNP 4	p28137-rs12229555	Intron 1	GG/AG/AA	G, 0.2188	31/207/377	0.0080	0.5310		25/135/271	-0.3159	0.5476	
12	SNP 1	p22116-rs1230382	Intron 1	GG/GA/AA	A, 0.4005	215/308/93	-0.0031	0.7775	**0.0178**	143/215/73	-0.0729	0.8730	0.9072
12	SNP 2	p26525-rs10773109	Intron 1	GG/CG/CC	G, 0.4176	102/312/203	-0.0132	0.2286		67/211/154	0.0192	0.9667	
12	SNP 3	p28137-rs12229555	Intron 1	GG/AG/AA	G, 0.2188	31/207/377	0.0080	0.5310		25/135/271	-0.3159	0.5476	
12	SNP 4	p28692-rs4765622	Intron 1	TT/CT/CC	T, 0.4693	136/307/173	0.0130	0.2255		109/209/113	-0.0560	0.8988	
13	SNP 1	p26525-rs10773109	Intron 1	GG/CG/CC	G, 0.4176	102/312/203	-0.0132	0.2286	0.0869	67/211/154	0.0192	0.9667	0.8434
13	SNP 2	p28137-rs12229555	Intron 1	GG/AG/AA	G, 0.2188	31/207/377	0.0080	0.5310		25/135/271	-0.3159	0.5476	
13	SNP 3	p28692-rs4765622	Intron 1	TT/CT/CC	T, 0.4693	136/307/173	0.0130	0.2255		109/209/113	-0.0560	0.8988	
13	SNP 4	p28957-rs11057844	Intron 1	GG/GA/AA	A, 0.1839	413/182/23	-0.0395	0.0035		302/116/14	0.2856	0.6271	
14	SNP 1	p28137-rs12229555	Intron 1	GG/AG/AA	G, 0.2188	31/207/377	0.0080	0.5310	**0.0181**	25/135/271	-0.3159	0.5476	0.6169
14	SNP 2	p28692-rs4765622	Intron 1	TT/CT/CC	T, 0.4693	136/307/173	0.0130	0.2255		109/209/113	-0.0560	0.8988	
14	SNP 3	p28957-rs11057844	Intron 1	GG/GA/AA	A, 0.1839	413/182/23	-0.0395	0.0035		302/116/14	0.2856	0.6271	

14	SNP 4	p31072-rs10846749	Intron 1	CC/CG/GG	G, 0.3448	266/269/75	0.0059	0.5976		189/185/51	0.0045	0.9922	
15	SNP 1	p28692-rs4765622	Intron 1	TT/CT/CC	T, 0.4693	136/307/173	0.0130	0.2255	**0.0068**	109/209/113	-0.0560	0.8988	0.6083
15	SNP 2	p28957-rs11057844	Intron 1	GG/GA/AA	A, 0.1839	413/182/23	-0.0395	0.0035		302/116/14	0.2856	0.6271	
15	SNP 3	p31072-rs10846749	Intron 1	CC/CG/GG	G, 0.3448	266/269/75	0.0059	0.5976		189/185/51	0.0045	0.9922	
15	SNP 4	p31938-rs10744182	Intron 1	GG/AC/AA	G, 0.4130	114/280/219	-0.0088	0.4065		74/201/155	-0.3933	0.3813	
16	SNP 1	p28957-rs11057844	Intron 1	GG/GA/AA	A, 0.1839	413/182/23	-0.0395	0.0035	**0.0092**	302/116/14	0.2856	0.6271	0.7179
16	SNP 2	p31072-rs10846749	Intron 1	CC/CG/GG	G, 0.3448	266/269/75	0.0059	0.5976		189/185/51	0.0045	0.9922	
16	SNP 3	p31938-rs10744182	Intron 1	GG/AC/AA	G, 0.4130	114/280/219	-0.0088	0.4065		74/201/155	-0.3933	0.3813	
16	SNP 4	p32129-rs10773107	Intron 1	TT/GT/GG	T, 0.4828	132/324/154	-0.0140	0.2065		97/219/111	0.5435	0.2270	
17	SNP 1	p31072-rs10846749	Intron 1	CC/CG/GG	G, 0.3448	266/269/75	0.0059	0.5976	0.3442	189/185/51	0.0045	0.9922	0.7354
17	SNP 2	p31938-rs10744182	Intron 1	GG/AC/AA	G, 0.4130	114/280/219	-0.0088	0.4065		74/201/155	-0.3933	0.3813	
17	SNP 3	p32129-rs10773107	Intron 1	TT/GT/GG	T, 0.4828	132/324/154	-0.0140	0.2065		97/219/111	0.5435	0.2270	
17	SNP 4	p32273-rs12580803	Intron 1	TT/TC/CC	C, 0.1726	418/181/16	0.0045	0.7527		305/111/13	0.0036	0.9952	
18	SNP 1	p31938-rs10744182	Intron 1	GG/AC/AA	G, 0.4130	114/280/219	-0.0088	0.4065	0.6301	74/201/155	-0.3933	0.3813	0.8332
18	SNP 2	p32129-rs10773107	Intron 1	TT/GT/GG	T, 0.4828	132/324/154	-0.0140	0.2065		97/219/111	0.5435	0.2270	
18	SNP 3	p32273-rs12580803	Intron 1	TT/TC/CC	C, 0.1726	418/181/16	0.0045	0.7527		305/111/13	0.0036	0.9952	
18	SNP 4	p32395-rs12581963	Intron 1	TT/CT/CC	T, 0.0829	5/93/521	0.0129	0.4973		4/67/362	-0.8103	0.2986	
19	SNP 1	p32129-rs10773107	Intron 1	TT/GT/GG	T, 0.4828	132/324/154	-0.0140	0.2065	0.2289	97/219/111	0.5435	0.2270	0.7201
19	SNP 2	p32273-rs12580803	Intron 1	TT/TC/CC	C, 0.1726	418/181/16	0.0045	0.7527		305/111/13	0.0036	0.9952	
19	SNP 3	p32395-rs12581963	Intron 1	TT/CT/CC	T, 0.0829	5/93/521	0.0129	0.4973		4/67/362	-0.8103	0.2986	
19	SNP 4	p32860-rs7967406	Intron 1	CC/AC/AA	C, 0.3921	96/291/232	0.0023	0.8303		75/206/151	0.7558	0.0920	
20	SNP 1	p32273-rs12580803	Intron 1	TT/TC/CC	C, 0.1726	418/181/16	0.0045	0.7527	0.2513	305/111/13	0.0036	0.9952	0.0944
20	SNP 2	p32395-rs12581963	Intron 1	TT/CT/CC	T, 0.0829	5/93/521	0.0129	0.4973		4/67/362	-0.8103	0.2986	
20	SNP 3	p32860-rs7967406	Intron 1	CC/AC/AA	C, 0.3921	96/291/232	0.0023	0.8303		75/206/151	0.7558	0.0920	
20	SNP 4	p36908-rs10846745	Intron 1	CC/CG/GG	G, 0.4748	169/305/140	0.0051	0.6330		130/212/87	-0.8517	0.0551	
21	SNP 1	p32395-rs12581963	Intron 1	TT/CT/CC	T, 0.0829	5/93/521	0.0129	0.4973	0.2764	4/67/362	-0.8103	0.2986	**0.0363**
21	SNP 2	p32860-rs7967406	Intron 1	CC/AC/AA	C, 0.3921	96/291/232	0.0023	0.8303		75/206/151	0.7558	0.0920	
21	SNP 3	p36908-rs10846745	Intron 1	CC/CG/GG	G, 0.4748	169/305/140	0.0051	0.6330		130/212/87	-0.8517	0.0551	
21	SNP 4	p37095-rs10846744	Intron 1	CC/CG/GG	G, 0.1364	458/144/12	-0.0132	0.3943		321/98/10	0.0774	0.9027	
22	SNP 1	p32860-rs7967406	Intron 1	CC/AC/AA	C, 0.3921	96/291/232	0.0023	0.8303	0.5791	75/206/151	0.7558	0.0920	0.1995
22	SNP 2	p36908-rs10846745	Intron 1	CC/CG/GG	G, 0.4748	169/305/140	0.0051	0.6330		130/212/87	-0.8517	0.0551	
22	SNP 3	p37095-rs10846744	Intron 1	CC/CG/GG	G, 0.1364	458/144/12	-0.0132	0.3943		321/98/10	0.0774	0.9027	
22	SNP 4	p42467-rs11057830	Intron 1	TT/CT/CC	T, 0.1329	12/140/463	-0.0120	0.4429		10/97/323	0.1439	0.8205	
23	SNP 1	p36908-rs10846745	Intron 1	CC/CG/GG	G, 0.4748	169/305/140	0.0051	0.6330	0.6904	130/212/87	-0.8517	0.0551	0.2715
23	SNP 2	p37095-rs10846744	Intron 1	CC/CG/GG	G, 0.1364	458/144/12	-0.0132	0.3943		321/98/10	0.0774	0.9027	
23	SNP 3	p42467-rs11057830	Intron 1	TT/CT/CC	T, 0.1329	12/140/463	-0.0120	0.4429		10/97/323	0.1439	0.8205	
23	SNP 4	p47235-rs199779577	Intron 1	CC/CT	T, 0.0016	613/2	0.1240	0.3520		428/1	2.1039	0.7471	
24	SNP 1	p37095-rs10846744	Intron 1	CC/CG/GG	G, 0.1364	458/144/12	-0.0132	0.3943	0.7036	321/98/10	0.0774	0.9027	0.9321
24	SNP 2	p42467-rs11057830	Intron 1	TT/CT/CC	T, 0.1329	12/140/463	-0.0120	0.4429		10/97/323	0.1439	0.8205	
24	SNP 3	p47235-rs199779577	Intron 1	CC/CT	T, 0.0016	613/2	0.1240	0.3520		428/1	2.1039	0.7471	
24	SNP 4	p47416-rs73227571	Exon 2	CC/CT	T, 0.0008	620/1	0.2718	0.1484		433/1	-12.3322	0.0599	
25	SNP 1	p42467-rs11057830	Intron 1	TT/CT/CC	T, 0.1329	12/140/463	-0.0120	0.4429	0.2721	10/97/323	0.1439	0.8205	**0.0341**
25	SNP 2	p47235-rs199779577	Intron 1	CC/CT	T, 0.0016	613/2	0.1240	0.3520		428/1	2.1039	0.7471	
25	SNP 3	p47416-rs73227571	Exon 2	CC/CT	T, 0.0008	620/1	0.2718	0.1484		433/1	-12.3322	0.0599	
25	SNP 4	p48969-rs2343394	Intron 2	TT/CT/CC	T, 0.2850	53/240/319	-0.0090	0.4412		41/174/213	1.2544	0.0082	
26	SNP 1	p47235-rs199779577	Intron 1	CC/CT	T, 0.0016	613/2	0.1240	0.3520	0.3710	428/1	2.1039	0.7471	**0.0293**
26	SNP 2	p47416-rs73227571	Exon 2	CC/CT	T, 0.0008	620/1	0.2718	0.1484		433/1	-12.3322	0.0599	
26	SNP 3	p48969-rs2343394	Intron 2	TT/CT/CC	T, 0.2850	53/240/319	-0.0090	0.4412		41/174/213	1.2544	0.0082	
26	SNP 4	p49518-rs144194221	Intron 2	GA/GG	A, 0.0217	27/593	0.0046	0.9017		20/413	1.4134	0.3456	
27	SNP 1	p47416-rs73227571	Exon 2	CC/CT	T, 0.0008	620/1	0.2718	0.1484	0.7122	433/1	-12.3322	0.0599	**0.0465**
27	SNP 2	p48969-rs2343394	Intron 2	TT/CT/CC	T, 0.2850	53/240/319	-0.0090	0.4412		41/174/213	1.2544	0.0082	
27	SNP 3	p49518-rs144194221	Intron 2	GA/GG	A, 0.0217	27/593	0.0046	0.9017		20/413	1.4134	0.3456	
27	SNP 4	p49690-rs4765615	Intron 2	GG/AG/AA	G, 0.4497	118/314/182	-0.0133	0.2263		90/220/120	1.2493	0.0059	
28	SNP 1	p48969-rs2343394	Intron 2	TT/CT/CC	T, 0.2850	53/240/319	-0.0090	0.4412	0.7084	41/174/213	1.2544	0.0082	**0.0005**
28	SNP 2	p49518-rs144194221	Intron 2	GA/GG	A, 0.0217	27/593	0.0046	0.9017		20/413	1.4134	0.3456	
28	SNP 3	p49690-rs4765615	Intron 2	GG/AG/AA	G, 0.4497	118/314/182	-0.0133	0.2263		90/220/120	1.2493	0.0059	
28	SNP 4	p49978-rs5891	Exon 3	GA/GG	A, 0.0096	12/609	0.0454	0.4043		8/426	5.8266	0.0120	
29	SNP 1	p49518-rs144194221	Intron 2	GA/GG	A, 0.0217	27/593	0.0046	0.9017	0.4558	20/413	1.4134	0.3456	**0.0031**
29	SNP 2	p49690-rs4765615	Intron 2	GG/AG/AA	G, 0.4497	118/314/182	-0.0133	0.2263		90/220/120	1.2493	0.0059	
29	SNP 3	p49978-rs5891	Exon 3	GA/GG	A, 0.0096	12/609	0.0454	0.4043		8/426	5.8266	0.0120	
29	SNP 4	p50151-rs2278986	Intron 3	CC/TC/TT	C, 0.2890	54/244/316	-0.0040	0.7308		42/175/211	1.1926	0.0122	
30	SNP 1	p49690-rs4765615	Intron 2	GG/AG/AA	G, 0.4497	118/314/182	-0.0133	0.2263	0.2599	90/220/120	1.2493	0.0059	**0.0021**
30	SNP 2	p49978-rs5891	Exon 3	GA/GG	A, 0.0096	12/609	0.0454	0.4043		8/426	5.8266	0.0120	

30	SNP 3	p50151-rs2278986	Intron 3	CC/TC/TT	C, 0.2890	54/244/316	-0.0040	0.7308		42/175/211	1.1926	0.0122	
30	SNP 4	p50432-chr12_125299088	Intron 3	CC/CT	T, 0.0040	614/5	0.0745	0.3762		429/3	0.2876	0.9396	
31	SNP 1	p49978-rs5891	Exon 3	GA/GG	A, 0.0096	12/609	0.0454	0.4043	0.5955	8/426	5.8266	0.0120	**0.0027**
31	SNP 2	p50151-rs2278986	Intron 3	CC/TC/TT	C, 0.2890	54/244/316	-0.0040	0.7308		42/175/211	1.1926	0.0122	
31	SNP 3	p50432-chr12_125299088	Intron 3	CC/CT	T, 0.0040	614/5	0.0745	0.3762		429/3	0.2876	0.9396	
31	SNP 4	p52556-rs11057820	Intron 4	GG/AG/AA	G, 0.4871	154/291/172	-0.0054	0.6003		114/203/115	0.8700	0.0436	
32	SNP 1	p50151-rs2278986	Intron 3	CC/TC/TT	C, 0.2890	54/244/316	-0.0040	0.7308	0.2706	42/175/211	1.1926	0.0122	0.0507
32	SNP 2	p50432-chr12_125299088	Intron 3	CC/CT	T, 0.0040	614/5	0.0745	0.3762		429/3	0.2876	0.9396	
32	SNP 3	p52556-rs11057820	Intron 4	GG/AG/AA	G, 0.4871	154/291/172	-0.0054	0.6003		114/203/115	0.8700	0.0436	
32	SNP 4	p53093-rs201977189	Exon 5	GA/GG	A, 0.0016	2/613	0.2937	0.0275		2/428	1.3306	0.7748	
33	SNP 1	p50432-chr12_125299088	Intron 3	CC/CT	T, 0.0040	614/5	0.0745	0.3762	0.4107	429/3	0.2876	0.9396	0.1268
33	SNP 2	p52556-rs11057820	Intron 4	GG/AG/AA	G, 0.4871	154/291/172	-0.0054	0.6003		114/203/115	0.8700	0.0436	
33	SNP 3	p53093-rs201977189	Exon 5	GA/GG	A, 0.0016	2/613	0.2937	0.0275		2/428	1.3306	0.7748	
33	SNP 4	p53248-rs140613481	Intron 5	TA/TT	A, 0.0008	1/611	-0.3175	0.0913		1/426	-0.6467	0.9209	
34	SNP 1	p52556-rs11057820	Intron 4	GG/AG/AA	G, 0.4871	154/291/172	-0.0054	0.6003	0.9251	114/203/115	0.8700	0.0436	0.0638
34	SNP 2	p53093-rs201977189	Exon 5	GA/GG	A, 0.0016	2/613	0.2937	0.0275		2/428	1.3306	0.7748	
34	SNP 3	p53248-rs140613481	Intron 5	TA/TT	A, 0.0008	1/611	-0.3175	0.0913		1/426	-0.6467	0.9209	
34	SNP 4	p54492-rs61762481	Intron 5	AA/GA/GG	A, 0.0468	3/52/562	0.0034	0.8882		3/39/388	-1.2096	0.2056	
35	SNP 1	p53093-rs201977189	Exon 5	GA/GG	A, 0.0016	2/613	0.2937	0.0275	0.2118	2/428	1.3306	0.7748	0.4022
35	SNP 2	p53248-rs140613481	Intron 5	TA/TT	A, 0.0008	1/611	-0.3175	0.0913		1/426	-0.6467	0.9209	
35	SNP 3	p54492-rs61762481	Intron 5	AA/GA/GG	A, 0.0468	3/52/562	0.0034	0.8882		3/39/388	-1.2096	0.2056	
35	SNP 4	p54866-chr12_125294654	Intron 6	GA/GG	A, 0.0008	1/616	0.3998	0.0332		1/429	-4.9002	0.4519	
36	SNP 1	p53248-rs140613481	Intron 5	TA/TT	A, 0.0008	1/611	-0.3175	0.0913	0.4716	1/426	-0.6467	0.9209	0.2445
36	SNP 2	p54492-rs61762481	Intron 5	AA/GA/GG	A, 0.0468	3/52/562	0.0034	0.8882		3/39/388	-1.2096	0.2056	
36	SNP 3	p54866-chr12_125294654	Intron 6	GA/GG	A, 0.0008	1/616	0.3998	0.0332		1/429	-4.9002	0.4519	
36	SNP 4	p55963-rs7134858	Intron 6	TT/CT/CC	T, 0.0759	6/80/531	-0.0235	0.2305		6/61/365	0.6119	0.4247	
37	SNP 1	p54492-rs61762481	Intron 5	AA/GA/GG	A, 0.0468	3/52/562	0.0034	0.8882	0.4655	3/39/388	-1.2096	0.2056	0.1789
37	SNP 2	p54866-chr12_125294654	Intron 6	GA/GG	A, 0.0008	1/616	0.3998	0.0332		1/429	-4.9002	0.4519	
37	SNP 3	p55963-rs7134858	Intron 6	TT/CT/CC	T, 0.0759	6/80/531	-0.0235	0.2305		6/61/365	0.6119	0.4247	
37	SNP 4	p56845-rs838902	Intron 6	AA/AG	G, 0.0211	587/26	0.0059	0.8737		412/18	-2.7951	0.0757	
38	SNP 1	p54866-chr12_125294654	Intron 6	GA/GG	A, 0.0008	1/616	0.3998	0.0332	0.6260	1/429	-4.9002	0.4519	0.1372
38	SNP 2	p55963-rs7134858	Intron 6	TT/CT/CC	T, 0.0759	6/80/531	-0.0235	0.2305		6/61/365	0.6119	0.4247	
38	SNP 3	p56845-rs838902	Intron 6	AA/AG	G, 0.0211	587/26	0.0059	0.8737		412/18	-2.7951	0.0757	
38	SNP 4	p57308-chr12_125292212	Intron 7	CC/CT	T, 0.0016	613/2	-0.0994	0.4547		428/1	-4.3898	0.4980	
39	SNP 1	p55963-rs7134858	Intron 6	TT/CT/CC	T, 0.0759	6/80/531	-0.0235	0.2305	0.7153	6/61/365	0.6119	0.4247	0.2297
39	SNP 2	p56845-rs838902	Intron 6	AA/AG	G, 0.0211	587/26	0.0059	0.8737		412/18	-2.7951	0.0757	
39	SNP 3	p57308-chr12_125292212	Intron 7	CC/CT	T, 0.0016	613/2	-0.0994	0.4547		428/1	-4.3898	0.4980	
39	SNP 4	p57508-rs71458866	Intron 7	GA/GG	A, 0.0162	20/594	-0.0043	0.9191		14/415	-3.1506	0.0723	
40	SNP 1	p56845-rs838902	Intron 6	AA/AG	G, 0.0211	587/26	0.0059	0.8737	0.9927	412/18	-2.7951	0.0757	0.1566
40	SNP 2	p57308-chr12_125292212	Intron 7	CC/CT	T, 0.0016	613/2	-0.0994	0.4547		428/1	-4.3898	0.4980	
40	SNP 3	p57508-rs71458866	Intron 7	GA/GG	A, 0.0162	20/594	-0.0043	0.9191		14/415	-3.1506	0.0723	
40	SNP 4	p57592-rs838903	Intron 7	GA/GG	A, 0.0161	20/598	-0.0045	0.9163		14/418	-3.1378	0.0758	
41	SNP 1	p57308-chr12_125292212	Intron 7	CC/CT	T, 0.0016	613/2	-0.0994	0.4547	0.8628	428/1	-4.3898	0.4980	0.1539
41	SNP 2	p57508-rs71458866	Intron 7	GA/GG	A, 0.0162	20/594	-0.0043	0.9191		14/415	-3.1506	0.0723	
41	SNP 3	p57592-rs838903	Intron 7	GA/GG	A, 0.0161	20/598	-0.0045	0.9163		14/418	-3.1378	0.0758	
41	SNP 4	p57618-chr12_125291902	Intron 7	GA/GG	A, 0.0008	1/615	0.3727	0.0470		NA	NA	NA	
42	SNP 1	p57508-rs71458866	Intron 7	GA/GG	A, 0.0162	20/594	-0.0043	0.9191	0.1990	14/415	-3.1506	0.0723	0.1575
42	SNP 2	p57592-rs838903	Intron 7	GA/GG	A, 0.0161	20/598	-0.0045	0.9163		14/418	-3.1378	0.0758	
42	SNP 3	p57618-chr12_125291902	Intron 7	GA/GG	A, 0.0008	1/615	0.3727	0.0470		NA	NA	NA	
42	SNP 4	p58514-rs838905	Intron 7	CT/TT	C, 0.0347	43/575	0.0372	0.2089		30/402	-2.0859	0.0914	
43	SNP 1	p57592-rs838903	Intron 7	GA/GG	A, 0.0161	20/598	-0.0045	0.9163	0.2371	14/418	-3.1378	0.0758	0.2305
43	SNP 2	p57618-chr12_125291902	Intron 7	GA/GG	A, 0.0008	1/615	0.3727	0.0470		NA	NA	NA	
43	SNP 3	p58514-rs838905	Intron 7	CT/TT	T, 0.0347	43/575	0.0372	0.2089		30/402	-2.0859	0.0914	
43	SNP 4	p60255-rs3782287	Intron 7	CC/CT/TT	T, 0.4766	174/296/147	-0.0122	0.2448		123/209/100	-0.2158	0.6229	
44	SNP 1	p57618-chr12_125291902	Intron 7	GA/GG	A, 0.0008	1/615	0.3727	0.0470	0.4486	NA	NA	NA	**0.0459**
44	SNP 2	p58514-rs838905	Intron 7	CT/TT	T, 0.0347	43/575	0.0372	0.2089		30/402	-2.0859	0.0914	
44	SNP 3	p60255-rs3782287	Intron 7	CC/CT/TT	T, 0.4766	174/296/147	-0.0122	0.2448		123/209/100	-0.2158	0.6229	
44	SNP 4	p63483-rs838912	Intron 7	AA/GA/GG	A, 0.4927	154/293/165	0.0125	0.2308		111/205/112	0.3773	0.3856	
45	SNP 1	p58514-rs838905	Intron 7	CT/TT	T, 0.0347	43/575	0.0372	0.2089	0.4980	30/402	-2.0859	0.0914	0.1002
45	SNP 2	p60255-rs3782287	Intron 7	CC/CT/TT	T, 0.4766	174/296/147	-0.0122	0.2448		123/209/100	-0.2158	0.6229	
45	SNP 3	p63483-rs838912	Intron 7	AA/GA/GG	A, 0.4927	154/293/165	0.0125	0.2308		111/205/112	0.3773	0.3856	
45	SNP 4	p64285-chr12_125285235	Intron 7	CC/CG	G, 0.0008	613/1	-0.3643	0.0525		428/1	-8.5632	0.1866	
46	SNP 1	p60255-rs3782287	Intron 7	CC/CT/TT	T, 0.4766	174/296/147	-0.0122	0.2448	0.6914	123/209/100	-0.2158	0.6229	0.6933

46	SNP 2	p63483-rs838912	Intron 7	AA/GA/GG	A, 0.4927	154/293/165	0.0125	0.2308		111/205/112	0.3773	0.3856	
46	SNP 3	p64285-chr12_125285235	Intron 7	CC/CG	G, 0.0008	613/1	-0.3643	0.0525		428/1	-8.5632	0.1866	
46	SNP 4	p64772-rs5888	Exon 8	CC/CT/TT	T, 0.4846	169/296/148	0.0068	0.5213		117/205/106	0.3416	0.4307	
47	SNP 1	p63483-rs838912	Intron 7	AA/GA/GG	A, 0.4927	154/293/165	0.0125	0.2308	0.6329	111/205/112	0.3773	0.3856	0.6536
47	SNP 2	p64285-chr12_125285235	Intron 7	CC/CG	G, 0.0008	613/1	-0.3643	0.0525		428/1	-8.5632	0.1866	
47	SNP 3	p64772-rs5888	Exon 8	CC/CT/TT	T, 0.4846	169/296/148	0.0068	0.5213		117/205/106	0.3416	0.4307	
47	SNP 4	p69612-rs147238482	Intron 8	AG/GG	A, 0.0048	6/615	-0.0154	0.8408		5/429	4.2242	0.1507	
48	SNP 1	p64285-chr12_125285235	Intron 7	CC/CG	G, 0.0008	613/1	-0.3643	0.0525	0.3052	428/1	-8.5632	0.1866	0.4601
48	SNP 2	p64772-rs5888	Exon 8	CC/CT/TT	T, 0.4846	169/296/148	0.0068	0.5213		117/205/106	0.3416	0.4307	
48	SNP 3	p69612-rs147238482	Intron 8	AG/GG	A, 0.0048	6/615	-0.0154	0.8408		5/429	4.2242	0.1507	
48	SNP 4	p72551-rs1031605	Intron 9	CC/CT/TT	T, 0.1755	419/182/18	0.0252	0.0731		297/122/14	0.4528	0.4378	
49	SNP 1	p64772-rs5888	Exon 8	CC/CT/TT	T, 0.4846	169/296/148	0.0068	0.5213	0.3779	117/205/106	0.3416	0.4307	0.2998
49	SNP 2	p69612-rs147238482	Intron 8	AG/GG	A, 0.0048	6/615	-0.0154	0.8408		5/429	4.2242	0.1507	
49	SNP 3	p72551-rs1031605	Intron 9	CC/CT/TT	T, 0.1755	419/182/18	0.0252	0.0731		297/122/14	0.4528	0.4378	
49	SNP 4	p76757-rs9919713	Intron 9	AA/AT/TT	T, 0.0379	573/43/2	0.0339	0.2127		399/31/1	-0.4687	0.6818	
50	SNP 1	p69612-rs147238482	Intron 8	AG/GG	A, 0.0048	6/615	-0.0154	0.8408	0.2703	5/429	4.2242	0.1507	0.2919
50	SNP 2	p72551-rs1031605	Intron 9	CC/CT/TT	T, 0.1755	419/182/18	0.0252	0.0731		297/122/14	0.4528	0.4378	
50	SNP 3	p76757-rs9919713	Intron 9	AA/AT/TT	T, 0.0379	573/43/2	0.0339	0.2127		399/31/1	-0.4687	0.6818	
50	SNP 4	p77250-rs201901986	Intron 9	GG/GT	T, 0.0225	592/28	0.0297	0.4112		413/20	-1.1574	0.4385	
51	SNP 1	p72551-rs1031605	Intron 9	CC/CT/TT	T, 0.1755	419/182/18	0.0252	0.0731	0.2857	297/122/14	0.4528	0.4378	0.7953
51	SNP 2	p76757-rs9919713	Intron 9	AA/AT/TT	T, 0.0379	573/43/2	0.0339	0.2127		399/31/1	-0.4687	0.6818	
51	SNP 3	p77250-rs201901986	Intron 9	GG/GT	T, 0.0225	592/28	0.0297	0.4112		413/20	-1.1574	0.4385	
51	SNP 4	p77251-rs34339961	Intron 9	AA/AT	T, 0.0226	590/28	0.0296	0.4144		412/20	-1.1124	0.4559	
52	SNP 1	p76757-rs9919713	Intron 9	AA/AT/TT	T, 0.0379	573/43/2	0.0339	0.2127	0.4458	399/31/1	-0.4687	0.6818	0.7283
52	SNP 2	p77250-rs201901986	Intron 9	GG/GT	T, 0.0225	592/28	0.0297	0.4112		413/20	-1.1574	0.4385	
52	SNP 3	p77251-rs34339961	Intron 9	AA/AT	T, 0.0226	590/28	0.0296	0.4144		412/20	-1.1124	0.4559	
52	SNP 4	p77842-rs2272310	Intron 10	AA/GA/GG	A, 0.0146	1/16/597	0.0414	0.3296		1/11/416	0.3875	0.8187	
53	SNP 1	p77250-rs201901986	Intron 9	GG/GT	T, 0.0225	592/28	0.0297	0.4112	0.2169	413/20	-1.1574	0.4385	**0.0075**
53	SNP 2	p77251-rs34339961	Intron 9	AA/AT	T, 0.0226	590/28	0.0296	0.4144		412/20	-1.1124	0.4559	
53	SNP 3	p77842-rs2272310	Intron 10	AA/GA/GG	A, 0.0146	1/16/597	0.0414	0.3296		1/11/416	0.3875	0.8187	
53	SNP 4	p78334-chr12_125271186	Intron 10	TG/TT	G, 0.0024	3/618	-0.1888	0.0811		2/432	14.5804	0.0015	
54	SNP 1	p77251-rs34339961	Intron 9	AA/AT	T, 0.0226	590/28	0.0296	0.4144	0.0768	412/20	-1.1124	0.4559	**0.0487**
54	SNP 2	p77842-rs2272310	Intron 10	AA/GA/GG	A, 0.0146	1/16/597	0.0414	0.3296		1/11/416	0.3875	0.8187	
54	SNP 3	p78334-chr12_125271186	Intron 10	TG/TT	G, 0.0024	3/618	-0.1888	0.0811		2/432	14.5804	0.0015	
54	SNP 4	p78430-rs838897	Intron 10	GG/CG/CC	G, 0.0364	2/41/573	0.0355	0.1996		1/28/401	-0.6962	0.5598	
55	SNP 1	p77842-rs2272310	Intron 10	AA/GA/GG	A, 0.0146	1/16/597	0.0414	0.3296	0.2288	1/11/416	0.3875	0.8187	0.1031
55	SNP 2	p78334-chr12_125271186	Intron 10	TG/TT	G, 0.0024	3/618	-0.1888	0.0811		2/432	14.5804	0.0015	
55	SNP 3	p78430-rs838897	Intron 10	GG/CG/CC	G, 0.0364	2/41/573	0.0355	0.1996		1/28/401	-0.6962	0.5598	
55	SNP 4	p78747-rs2293440	Intron 11	CC/TC/TT	C, 0.0243	1/28/587	0.0378	0.2647		1/18/410	-0.8158	0.5651	
56	SNP 1	p78334-chr12_125271186	Intron 10	TG/TT	G, 0.0024	3/618	-0.1888	0.0811	0.6323	2/432	14.5804	0.0015	0.3441
56	SNP 2	p78430-rs838897	Intron 10	GG/CG/CC	G, 0.0364	2/41/573	0.0355	0.1996		1/28/401	-0.6962	0.5598	
56	SNP 3	p78747-rs2293440	Intron 11	CC/TC/TT	C, 0.0243	1/28/587	0.0378	0.2647		1/18/410	-0.8158	0.5651	
56	SNP 4	p79721-rs838896	Intron 11	GG/GC/CC	C, 0.3169	292/257/66	0.0086	0.4506		204/173/52	0.6682	0.1436	
57	SNP 1	p78430-rs838897	Intron 10	GG/CG/CC	G, 0.0364	2/41/573	0.0355	0.1996	0.4552	1/28/401	-0.6962	0.5598	0.7514
57	SNP 2	p78747-rs2293440	Intron 11	CC/TC/TT	C, 0.0243	1/28/587	0.0378	0.2647		1/18/410	-0.8158	0.5651	
57	SNP 3	p79721-rs838896	Intron 11	GG/GC/CC	C, 0.3169	292/257/66	0.0086	0.4506		204/173/52	0.6682	0.1436	
57	SNP 4	p79828-rs838895	Intron 11	GG/CG/CC	G, 0.2891	53/243/309	0.0169	0.1489		41/170/212	0.5448	0.2495	
58	SNP 1	p78747-rs2293440	Intron 11	CC/TC/TT	C, 0.0243	1/28/587	0.0378	0.2647	0.6932	1/18/410	-0.8158	0.5651	0.5529
58	SNP 2	p79721-rs838896	Intron 11	GG/GC/CC	C, 0.3169	292/257/66	0.0086	0.4506		204/173/52	0.6682	0.1436	
58	SNP 3	p79828-rs838895	Intron 11	GG/CG/CC	G, 0.2891	53/243/309	0.0169	0.1489		41/170/212	0.5448	0.2495	
58	SNP 4	p80045-rs838893	Intron 11	GG/GA/AA	A, 0.2879	315/251/52	0.0136	0.2472		216/175/40	0.7795	0.1050	
59	SNP 1	p79721-rs838896	Intron 11	GG/GC/CC	C, 0.3169	292/257/66	0.0086	0.4506	0.6338	204/173/52	0.6682	0.1436	0.5492
59	SNP 2	p79828-rs838895	Intron 11	GG/CG/CC	G, 0.2891	53/243/309	0.0169	0.1489		41/170/212	0.5448	0.2495	
59	SNP 3	p80045-rs838893	Intron 11	GG/GA/AA	A, 0.2879	315/251/52	0.0136	0.2472		216/175/40	0.7795	0.1050	
59	SNP 4	p83088-rs7977729	Intron 12	GG/AG/AA	G, 0.2669	44/242/332	0.0107	0.3768		34/167/230	0.7858	0.1140	
60	SNP 1	p79828-rs838895	Intron 11	GG/CG/CC	G, 0.2891	53/243/309	0.0169	0.1489	**0.0453**	41/170/212	0.5448	0.2495	0.2618
60	SNP 2	p80045-rs838893	Intron 11	GG/GA/AA	A, 0.2879	315/251/52	0.0136	0.2472		216/175/40	0.7795	0.1050	
60	SNP 3	p83088-rs7977729	Intron 12	GG/AG/AA	G, 0.2669	44/242/332	0.0107	0.3768		34/167/230	0.7858	0.1140	
60	SNP 4	p83884-rs701106	Intron 12	TT/CT/CC	T, 0.1527	17/155/448	0.0394	0.0066		12/116/305	-0.3865	0.5179	
61	SNP 1	p80045-rs838893	Intron 11	GG/GA/AA	A, 0.2879	315/251/52	0.0136	0.2472	**0.0236**	216/175/40	0.7795	0.1050	0.2843
61	SNP 2	p83088-rs7977729	Intron 12	GG/AG/AA	G, 0.2669	44/242/332	0.0107	0.3768		34/167/230	0.7858	0.1140	
61	SNP 3	p83884-rs701106	Intron 12	TT/CT/CC	T, 0.1527	17/155/448	0.0394	0.0066		12/116/305	-0.3865	0.5179	
61	SNP 4	p86436-rs10396214	Exon 13-3' UTR	CC/CT	T, 0.0088	610/11	-0.0402	0.4808		425/9	-0.6137	0.7810	

62	SNP 1	p83088-rs7977729	Intron 12	GG/AG/AA	G, 0.2669	44/242/332	0.0107	0.3768	**0.0034**	34/167/230	0.7858	0.1140	0.2740
62	SNP 2	p83884-rs701106	Intron 12	TT/CT/CC	T, 0.1527	17/155/448	0.0394	0.0066		12/116/305	-0.3865	0.5179	
62	SNP 3	p86436-rs10396214	Exon 13-3' UTR	CC/CT	T, 0.0088	610/11	-0.0402	0.4808		425/9	-0.6137	0.7810	
62	SNP 4	p87004-rs184715678	Exon 13-3' UTR	CA/CC	A, 0.0016	2/617	-0.2166	0.1019		2/430	2.9213	0.5277	
63	SNP 1	p83884-rs701106	Intron 12	TT/CT/CC	T, 0.1527	17/155/448	0.0394	0.0066	**0.0047**	12/116/305	-0.3865	0.5179	0.4943
63	SNP 2	p86436-rs10396214	Exon 13-3'UTR	CC/CT	T, 0.0088	610/11	-0.0402	0.4808		425/9	-0.6137	0.7810	
63	SNP 3	p87004-rs184715678	Exon 13-3' UTR	CA/CC	A, 0.0016	2/617	-0.2166	0.1019		2/430	2.9213	0.5277	
63	SNP 4	p87210-chr12_125262310	Exon 13-3' UTR	GG/GA	A, 0.0016	619/2	-0.0914	0.4911		432/2	6.3910	0.1675	
64	SNP 1	p86436-rs10396214	Exon 13-3' UTR	CC/CT	T, 0.0088	610/11	-0.0402	0.4808	0.1580	425/9	-0.6137	0.7810	0.5124
64	SNP 2	p87004-rs184715678	Exon 13-3' UTR	CA/CC	A, 0.0016	2/617	-0.2166	0.1019		2/430	2.9213	0.5277	
64	SNP 3	p87210-chr12_125262310	Exon 13-3' UTR	GG/GA	A, 0.0016	619/2	-0.0914	0.4911		432/2	6.3910	0.1675	
64	SNP 4	p87381-rs192190977	3' flanking	CC/CT	T, 0.0024	616/3	0.0885	0.4149		429/3	4.2516	0.2608	
65	SNP 1	p87004-rs184715678	Exon 13-3' UTR	CA/CC	A, 0.0016	2/617	-0.2166	0.1019	0.5896	2/430	2.9213	0.5277	0.1217
65	SNP 2	p87210-chr12_125262310	Exon 13-3' UTR	GG/GA	A, 0.0016	619/2	-0.0914	0.4911		432/2	6.3910	0.1675	
65	SNP 3	p87381-rs192190977	3' flanking	CC/CT	T, 0.0024	616/3	0.0885	0.4149		429/3	4.2516	0.2608	
65	SNP 4	p87681-rs838883	3' flanking	GG/GA/AA	A, 0.0845	518/99/2	0.0152	0.4416		361/69/2	-0.8318	0.3044	
66	SNP 1	p87210-chr12_125262310	Exon 13-3' UTR	GG/GA	A, 0.0016	619/2	-0.0914	0.4911	0.1645	432/2	6.3910	0.1675	0.1557
66	SNP 2	p87381-rs192190977	3' flanking	CC/CT	T, 0.0024	616/3	0.0885	0.4149		429/3	4.2516	0.2608	
66	SNP 3	p87681-rs838883	3' flanking	GG/GA/AA	A, 0.0845	518/99/2	0.0152	0.4416		361/69/2	-0.8318	0.3044	
66	SNP 4	p87927-rs838880	3' flanking	AA/AG/GG	G, 0.3237	281/276/62	0.0257	0.0250		198/189/45	-0.7008	0.1398	

ApoB, apolipoprotein B; GT; genotype; HDL-C, high-density lipoprotein cholesterol; MA, minor allele; MAF, minor allele frequency; NA, not analyzed; SNP, single nucleotide polymorphism; UTR, untranslated region.
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. HDL-C and ApoB values were Box-Cox transformed. Results were adjusted for covariates (HDL-C: sex, age, smoking, and body mass index; ApoB: age and smoking).
Single-site *P*-values were unadjusted for multiple testing, see details in Tables A8 and A9.
Significant global *P*-values (<0.05) are shown in **bold**.
*Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
†dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).

Table A15. Haplotype association results of 69 *SCARB1* genotyped variants (haplotype windows and global *P*-values) for LDL-C and TG.

						LDL-C				TG			
wind #	wind SNP	SNP Name*-SNP ID†/ Chr12 Position*	Location	Genotype	MA, MAF	GT Count	β	Single site *P*	Global *P*	GT Count	β	Single site *P*	Global *P*
1	SNP 1	p1257-rs4238001	Exon 1	AA/GA/GG	A, 0.0818	5/91/521	-0.7889	0.8481	**0.0387**	5/90/518	-0.0201	0.6295	0.9196
1	SNP 2	p1588-chr12_125347932	Intron 1	GC/GG	C, 0.0008	1/613	-34.2394	0.3930		1/609	-0.1974	0.6262	
1	SNP 3	p4072-rs7139401	Intron 1	TT/CT/CC	T, 0.4564	136/293/190	1.2267	0.5771		135/291/190	-0.0140	0.5353	
1	SNP 4	p5055-rs11057869	Intron 1	GG/GA/AA	A, 0.0875	514/98/5	-2.4528	0.5418		511/97/5	-0.0162	0.6914	
2	SNP 1	p1588-chr12_125347932	Intron 1	GC/GG	C, 0.0008	1/613	-34.2394	0.3930	0.5090	1/609	-0.1974	0.6262	0.9530
2	SNP 2	p4072-rs7139401	Intron 1	TT/CT/CC	T, 0.4564	136/293/190	1.2267	0.5771		135/291/190	-0.0140	0.5353	
2	SNP 3	p5055-rs11057869	Intron 1	GG/GA/AA	A, 0.0875	514/98/5	-2.4528	0.5418		511/97/5	-0.0162	0.6914	
2	SNP 4	p7650-rs11615630	Intron 1	GG/GA/AA	A, 0.3817	242/279/96	3.1377	0.1608		241/278/96	-0.0046	0.8428	
3	SNP 1	p4072-rs7139401	Intron 1	TT/CT/CC	T, 0.4564	136/293/190	1.2267	0.5771	0.6333	135/291/190	-0.0140	0.5353	0.8981
3	SNP 2	p5055-rs11057869	Intron 1	GG/GA/AA	A, 0.0875	514/98/5	-2.4528	0.5418		511/97/5	-0.0162	0.6914	
3	SNP 3	p7650-rs11615630	Intron 1	GG/GA/AA	A, 0.3817	242/279/96	3.1377	0.1608		241/278/96	-0.0046	0.8428	
3	SNP 4	p10292-rs4765181	Intron 1	TT/GT/GG	T, 0.3972	94/303/221	-0.6380	0.7845		94/301/220	0.0312	0.1884	
4	SNP 1	p5055-rs11057869	Intron 1	GG/GA/AA	A, 0.0875	514/98/5	-2.4528	0.5418	0.7087	511/97/5	-0.0162	0.6914	0.6731
4	SNP 2	p7650-rs11615630	Intron 1	GG/GA/AA	A, 0.3817	242/279/96	3.1377	0.1608		241/278/96	-0.0046	0.8428	
4	SNP 3	p10292-rs4765181	Intron 1	TT/GT/GG	T, 0.3972	94/303/221	-0.6380	0.7845		94/301/220	0.0312	0.1884	
4	SNP 4	p13570-rs11057864	Intron 1	GT/GG	T, 0.0427	53/568	-1.9860	0.7274		53/565	-0.0300	0.6046	
5	SNP 1	p7650-rs11615630	Intron 1	GG/GA/AA	A, 0.3817	242/279/96	3.1377	0.1608	0.5153	241/278/96	-0.0046	0.8428	0.2204
5	SNP 2	p10292-rs4765181	Intron 1	TT/GT/GG	T, 0.3972	94/303/221	-0.6380	0.7845		94/301/220	0.0312	0.1884	
5	SNP 3	p13570-rs11057864	Intron 1	GT/GG	T, 0.0427	53/568	-1.9860	0.7274		53/565	-0.0300	0.6046	
5	SNP 4	p16565-rs10773111	Intron 1	GG/GA/AA	A, 0.4249	203/306/110	-2.8406	0.2153		203/303/110	0.0087	0.7098	
6	SNP 1	p10292-rs4765181	Intron 1	TT/GT/GG	T, 0.3972	94/303/221	-0.6380	0.7845	0.2616	94/301/220	0.0312	0.1884	0.5240
6	SNP 2	p13570-rs11057864	Intron 1	GT/GG	T, 0.0427	53/568	-1.9860	0.7274		53/565	-0.0300	0.6046	
6	SNP 3	p16565-rs10773111	Intron 1	GG/GA/AA	A, 0.4249	203/306/110	-2.8406	0.2153		203/303/110	0.0087	0.7098	
6	SNP 4	p20207-rs11057853	Intron 1	GG/AG/AA	G, 0.4269	109/308/199	2.4118	0.2953		109/306/198	-0.0016	0.9468	
7	SNP 1	p13570-rs11057864	Intron 1	GT/GG	T, 0.0427	53/568	-1.9860	0.7274	0.3808	53/565	-0.0300	0.6046	0.6131
7	SNP 2	p16565-rs10773111	Intron 1	GG/GA/AA	A, 0.4249	203/306/110	-2.8406	0.2153		203/303/110	0.0087	0.7098	
7	SNP 3	p20207-rs11057853	Intron 1	GG/AG/AA	G, 0.4269	109/308/199	2.4118	0.2953		109/306/198	-0.0016	0.9468	
7	SNP 4	p20741-rs11057851	Intron 1	TT/CT/CC	T, 0.1157	9/125/484	-1.2196	0.7290		9/124/482	-0.0536	0.1357	
8	SNP 1	p16565-rs10773111	Intron 1	GG/GA/AA	A, 0.4249	203/306/110	-2.8406	0.2153	0.1674	203/303/110	0.0087	0.7098	0.7548
8	SNP 2	p20207-rs11057853	Intron 1	GG/AG/AA	G, 0.4269	109/308/199	2.4118	0.2953		109/306/198	-0.0016	0.9468	
8	SNP 3	p20741-rs11057851	Intron 1	TT/CT/CC	T, 0.1157	9/125/484	-1.2196	0.7290		9/124/482	-0.0536	0.1357	
8	SNP 4	p21145-rs3924313	Intron 1	TT/CT/CC	T, 0.3083	52/277/289	3.9825	0.1078		52/277/287	0.0249	0.3297	
9	SNP 1	p20207-rs11057853	Intron 1	GG/AG/AA	G, 0.4269	109/308/199	2.4118	0.2953	0.1136	109/306/198	-0.0016	0.9468	0.5726
9	SNP 2	p20741-rs11057851	Intron 1	TT/CT/CC	T, 0.1157	9/125/484	-1.2196	0.7290		9/124/482	-0.0536	0.1357	
9	SNP 3	p21145-rs3924313	Intron 1	TT/CT/CC	T, 0.3083	52/277/289	3.9825	0.1078		52/277/287	0.0249	0.3297	
9	SNP 4	p22116-rs1230382	Intron 1	GG/GA/AA	A, 0.4005	216/309/93	-2.6507	0.2634		216/305/93	-0.0060	0.8024	
10	SNP 1	p20741-rs11057851	Intron 1	TT/CT/CC	T, 0.1157	9/125/484	-1.2196	0.7290	0.1921	9/124/482	-0.0536	0.1357	0.7044
10	SNP 2	p21145-rs3924313	Intron 1	TT/CT/CC	T, 0.3083	52/277/289	3.9825	0.1078		52/277/287	0.0249	0.3297	
10	SNP 3	p22116-rs1230382	Intron 1	GG/GA/AA	A, 0.4005	216/309/93	-2.6507	0.2634		216/305/93	-0.0060	0.8024	
10	SNP 4	p26525-rs10773109	Intron 1	GG/CG/CC	G, 0.4176	102/313/204	1.8031	0.4434		102/310/203	-0.0027	0.9078	
11	SNP 1	p21145-rs3924313	Intron 1	TT/CT/CC	T, 0.3083	52/277/289	3.9825	0.1078	0.4326	52/277/287	0.0249	0.3297	0.9237
11	SNP 2	p22116-rs1230382	Intron 1	GG/GA/AA	A, 0.4005	216/309/93	-2.6507	0.2634		216/305/93	-0.0060	0.8024	
11	SNP 3	p26525-rs10773109	Intron 1	GG/CG/CC	G, 0.4176	102/313/204	1.8031	0.4434		102/310/203	-0.0027	0.9078	
11	SNP 4	p28137-rs12229555	Intron 1	GG/AG/AA	G, 0.2188	31/208/378	0.2612	0.9242		31/207/375	-0.0086	0.7556	
12	SNP 1	p22116-rs1230382	Intron 1	GG/GA/AA	A, 0.4005	216/309/93	-2.6507	0.2634	0.7645	216/305/93	-0.0060	0.8024	0.9667
12	SNP 2	p26525-rs10773109	Intron 1	GG/CG/CC	G, 0.4176	102/313/204	1.8031	0.4434		102/310/203	-0.0027	0.9078	
12	SNP 3	p28137-rs12229555	Intron 1	GG/AG/AA	G, 0.2188	31/208/378	0.2612	0.9242		31/207/375	-0.0086	0.7556	
12	SNP 4	p28692-rs4765622	Intron 1	TT/CT/CC	T, 0.4693	136/308/174	-1.8314	0.4241		136/304/174	-0.0014	0.9512	
13	SNP 1	p26525-rs10773109	Intron 1	GG/CG/CC	G, 0.4176	102/313/204	1.8031	0.4434	0.9532	102/310/203	-0.0027	0.9078	0.9900
13	SNP 2	p28137-rs12229555	Intron 1	GG/AG/AA	G, 0.2188	31/208/378	0.2612	0.9242		31/207/375	-0.0086	0.7556	
13	SNP 3	p28692-rs4765622	Intron 1	TT/CT/CC	T, 0.4693	136/308/174	-1.8314	0.4241		136/304/174	-0.0014	0.9512	
13	SNP 4	p28957-rs11057844	Intron 1	GG/GA/AA	A, 0.1839	415/182/23	2.0109	0.4846		414/180/23	-0.0071	0.8088	
14	SNP 1	p28137-rs12229555	Intron 1	GG/AG/AA	G, 0.2188	31/208/378	0.2612	0.9242	0.9242	31/207/375	-0.0086	0.7556	0.8628
14	SNP 2	p28692-rs4765622	Intron 1	TT/CT/CC	T, 0.4693	136/308/174	-1.8314	0.4241		136/304/174	-0.0014	0.9512	
14	SNP 3	p28957-rs11057844	Intron 1	GG/GA/AA	A, 0.1839	415/182/23	2.0109	0.4846		414/180/23	-0.0071	0.8088	

14	SNP 4	p31072-rs10846749	Intron 1	CC/CG/GG	G, 0.3448	266/270/76	1.0276	0.6630		264/269/76	0.0114	0.6390	
15	SNP 1	p28692-rs4765622	Intron 1	TT/CT/CC	T, 0.4693	136/308/174	-1.8314	0.4241	0.7621	136/304/174	-0.0014	0.9512	0.1331
15	SNP 2	p28957-rs11057844	Intron 1	GG/GA/AA	A, 0.1839	415/182/23	2.0109	0.4846		414/180/23	-0.0071	0.8088	
15	SNP 3	p31072-rs10846749	Intron 1	CC/CG/GG	G, 0.3448	266/270/76	1.0276	0.6630		264/269/76	0.0114	0.6390	
15	SNP 4	p31938-rs10744182	Intron 1	GG/AC/AA	G, 0.4130	114/280/221	0.8915	0.6875		114/279/220	0.0182	0.4238	
16	SNP 1	p28957-rs11057844	Intron 1	GG/GA/AA	A, 0.1839	415/182/23	2.0109	0.4846	0.6369	414/180/23	-0.0071	0.8088	**0.0315**
16	SNP 2	p31072-rs10846749	Intron 1	CC/CG/GG	G, 0.3448	266/270/76	1.0276	0.6630		264/269/76	0.0114	0.6390	
16	SNP 3	p31938-rs10744182	Intron 1	GG/AC/AA	G, 0.4130	114/280/221	0.8915	0.6875		114/279/220	0.0182	0.4238	
16	SNP 4	p32129-rs10773107	Intron 1	TT/GT/GG	T, 0.4828	133/325/154	-0.8334	0.7206		132/323/154	-0.0439	0.0665	
17	SNP 1	p31072-rs10846749	Intron 1	CC/CG/GG	G, 0.3448	266/270/76	1.0276	0.6630	0.9335	264/269/76	0.0114	0.6390	**0.0100**
17	SNP 2	p31938-rs10744182	Intron 1	GG/AC/AA	G, 0.4130	114/280/221	0.8915	0.6875		114/279/220	0.0182	0.4238	
17	SNP 3	p32129-rs10773107	Intron 1	TT/GT/GG	T, 0.4828	133/325/154	-0.8334	0.7206		132/323/154	-0.0439	0.0665	
17	SNP 4	p32273-rs12580803	Intron 1	TT/TC/CC	C, 0.1726	420/181/16	0.1430	0.9627		417/180/16	0.0539	0.0807	
18	SNP 1	p31938-rs10744182	Intron 1	GG/AC/AA	G, 0.4130	114/280/221	0.8915	0.6875	0.6646	114/279/220	0.0182	0.4238	0.4958
18	SNP 2	p32129-rs10773107	Intron 1	TT/GT/GG	T, 0.4828	133/325/154	-0.8334	0.7206		132/323/154	-0.0439	0.0665	
18	SNP 3	p32273-rs12580803	Intron 1	TT/TC/CC	C, 0.1726	420/181/16	0.1430	0.9627		417/180/16	0.0539	0.0807	
18	SNP 4	p32395-rs12581963	Intron 1	TT/CT/CC	T, 0.0829	5/93/523	1.0504	0.7953		5/92/521	0.0137	0.7398	
19	SNP 1	p32129-rs10773107	Intron 1	TT/GT/GG	T, 0.4828	133/325/154	-0.8334	0.7206	0.5135	132/323/154	-0.0439	0.0665	0.2857
19	SNP 2	p32273-rs12580803	Intron 1	TT/TC/CC	C, 0.1726	420/181/16	0.1430	0.9627		417/180/16	0.0539	0.0807	
19	SNP 3	p32395-rs12581963	Intron 1	TT/CT/CC	T, 0.0829	5/93/523	1.0504	0.7953		5/92/521	0.0137	0.7398	
19	SNP 4	p32860-rs7967406	Intron 1	CC/AC/AA	C, 0.3921	98/291/232	0.1418	0.9509		98/288/231	-0.0322	0.1668	
20	SNP 1	p32273-rs12580803	Intron 1	TT/TC/CC	C, 0.1726	420/181/16	0.1430	0.9627	0.9948	417/180/16	0.0539	0.0807	0.4421
20	SNP 2	p32395-rs12581963	Intron 1	TT/CT/CC	T, 0.0829	5/93/523	1.0504	0.7953		5/92/521	0.0137	0.7398	
20	SNP 3	p32860-rs7967406	Intron 1	CC/AC/AA	C, 0.3921	98/291/232	0.1418	0.9509		98/288/231	-0.0322	0.1668	
20	SNP 4	p36908-rs10846745	Intron 1	CC/CG/GG	G, 0.4748	171/305/140	-0.1277	0.9553		169/303/140	0.0206	0.3706	
21	SNP 1	p32395-rs12581963	Intron 1	TT/CT/CC	T, 0.0829	5/93/523	1.0504	0.7953	0.8006	5/92/521	0.0137	0.7398	0.7373
21	SNP 2	p32860-rs7967406	Intron 1	CC/AC/AA	C, 0.3921	98/291/232	0.1418	0.9509		98/288/231	-0.0322	0.1668	
21	SNP 3	p36908-rs10846745	Intron 1	CC/CG/GG	G, 0.4748	171/305/140	-0.1277	0.9553		169/303/140	0.0206	0.3706	
21	SNP 4	p37095-rs10846744	Intron 1	CC/CG/GG	G, 0.1364	460/144/12	-0.6209	0.8512		460/140/12	0.0170	0.6132	
22	SNP 1	p32860-rs7967406	Intron 1	CC/AC/AA	C, 0.3921	98/291/232	0.1418	0.9509	0.9985	98/288/231	-0.0322	0.1668	0.5690
22	SNP 2	p36908-rs10846745	Intron 1	CC/CG/GG	G, 0.4748	171/305/140	-0.1277	0.9553		169/303/140	0.0206	0.3706	
22	SNP 3	p37095-rs10846744	Intron 1	CC/CG/GG	G, 0.1364	460/144/12	-0.6209	0.8512		460/140/12	0.0170	0.6132	
22	SNP 4	p42467-rs11057830	Intron 1	TT/CT/CC	T, 0.1329	12/140/465	0.5145	0.8756		12/138/464	0.0207	0.5394	
23	SNP 1	p36908-rs10846745	Intron 1	CC/CG/GG	G, 0.4748	171/305/140	-0.1277	0.9553	0.9589	169/303/140	0.0206	0.3706	0.6256
23	SNP 2	p37095-rs10846744	Intron 1	CC/CG/GG	G, 0.1364	460/144/12	-0.6209	0.8512		460/140/12	0.0170	0.6132	
23	SNP 3	p42467-rs11057830	Intron 1	TT/CT/CC	T, 0.1329	12/140/465	0.5145	0.8756		12/138/464	0.0207	0.5394	
23	SNP 4	p47235-rs199779577	Intron 1	CC/CT	T, 0.0016	615/2	-7.4285	0.7935		611/2	0.1488	0.6034	
24	SNP 1	p37095-rs10846744	Intron 1	CC/CG/GG	G, 0.1364	460/144/12	-0.6209	0.8512	0.5091	460/140/12	0.0170	0.6132	0.8740
24	SNP 2	p42467-rs11057830	Intron 1	TT/CT/CC	T, 0.1329	12/140/465	0.5145	0.8756		12/138/464	0.0207	0.5394	
24	SNP 3	p47235-rs199779577	Intron 1	CC/CT	T, 0.0016	615/2	-7.4285	0.7935		611/2	0.1488	0.6034	
24	SNP 4	p47416-rs73227571	Exon 2	CC/CT	T, 0.0008	622/1	-94.8697	0.0179		618/1	-0.5814	0.1521	
25	SNP 1	p42467-rs11057830	Intron 1	TT/CT/CC	T, 0.1329	12/140/465	0.5145	0.8756	0.4595	12/138/464	0.0207	0.5394	0.9114
25	SNP 2	p47235-rs199779577	Intron 1	CC/CT	T, 0.0016	615/2	-7.4285	0.7935		611/2	0.1488	0.6034	
25	SNP 3	p47416-rs73227571	Exon 2	CC/CT	T, 0.0008	622/1	-94.8697	0.0179		618/1	-0.5814	0.1521	
25	SNP 4	p48969-rs2343394	Intron 2	TT/CT/CC	T, 0.2850	55/240/319	0.7714	0.7539		55/238/317	0.0031	0.9015	
26	SNP 1	p47235-rs199779577	Intron 1	CC/CT	T, 0.0016	615/2	-7.4285	0.7935	0.4646	611/2	0.1488	0.6034	0.9821
26	SNP 2	p47416-rs73227571	Exon 2	CC/CT	T, 0.0008	622/1	-94.8697	0.0179		618/1	-0.5814	0.1521	
26	SNP 3	p48969-rs2343394	Intron 2	TT/CT/CC	T, 0.2850	55/240/319	0.7714	0.7539		55/238/317	0.0031	0.9015	
26	SNP 4	p49518-rs144194221	Intron 2	GA/GG	A, 0.0217	27/595	-0.0995	0.9900		27/591	-0.0009	0.9913	
27	SNP 1	p47416-rs73227571	Exon 2	CC/CT	T, 0.0008	622/1	-94.8697	0.0179	0.6794	618/1	-0.5814	0.1521	0.9975
27	SNP 2	p48969-rs2343394	Intron 2	TT/CT/CC	T, 0.2850	55/240/319	0.7714	0.7539		55/238/317	0.0031	0.9015	
27	SNP 3	p49518-rs144194221	Intron 2	GA/GG	A, 0.0217	27/595	-0.0995	0.9900		27/591	-0.0009	0.9913	
27	SNP 4	p49690-rs4765615	Intron 2	GG/AG/AA	G, 0.4497	120/314/182	-0.7893	0.7348		118/313/181	0.0007	0.9779	
28	SNP 1	p48969-rs2343394	Intron 2	TT/CT/CC	T, 0.2850	55/240/319	0.7714	0.7539	0.7013	55/238/317	0.0031	0.9015	0.5651
28	SNP 2	p49518-rs144194221	Intron 2	GA/GG	A, 0.0217	27/595	-0.0995	0.9900		27/591	-0.0009	0.9913	
28	SNP 3	p49690-rs4765615	Intron 2	GG/AG/AA	G, 0.4497	120/314/182	-0.7893	0.7348		118/313/181	0.0007	0.9779	
28	SNP 4	p49978-rs5891	Exon 3	GA/GG	A, 0.0096	12/611	-13.3163	0.2525		12/607	-0.2536	0.0306	
29	SNP 1	p49518-rs144194221	Intron 2	GA/GG	A, 0.0217	27/595	-0.0995	0.9900	0.8775	27/591	-0.0009	0.9913	0.4377
29	SNP 2	p49690-rs4765615	Intron 2	GG/AG/AA	G, 0.4497	120/314/182	-0.7893	0.7348		118/313/181	0.0007	0.9779	
29	SNP 3	p49978-rs5891	Exon 3	GA/GG	A, 0.0096	12/611	-13.3163	0.2525		12/607	-0.2536	0.0306	
29	SNP 4	p50151-rs2278986	Intron 3	CC/TC/TT	C, 0.2890	56/244/316	0.0131	0.9957		56/242/314	0.0020	0.9377	
30	SNP 1	p49690-rs4765615	Intron 2	GG/AG/AA	G, 0.4497	120/314/182	-0.7893	0.7348	0.7111	118/313/181	0.0007	0.9779	0.3669
30	SNP 2	p49978-rs5891	Exon 3	GA/GG	A, 0.0096	12/611	-13.3163	0.2525		12/607	-0.2536	0.0306	

30	SNP 3	p50151-rs2278986	Intron 3	CC/TC/TT	C, 0.2890	56/244/316	0.0131	0.9957		56/242/314	0.0020	0.9377	
30	SNP 4	p50432-chr12_125299088	Intron 3	CC/CT	T, 0.0040	616/5	0.0726	0.9968		612/5	-0.0369	0.8387	
31	SNP 1	p49978-rs5891	Exon 3	GA/GG	A, 0.0096	12/611	-13.3163	0.2525	0.9754	12/607	-0.2536	0.0306	0.1828
31	SNP 2	p50151-rs2278986	Intron 3	CC/TC/TT	C, 0.2890	56/244/316	0.0131	0.9957		56/242/314	0.0020	0.9377	
31	SNP 3	p50432-chr12_125299088	Intron 3	CC/CT	T, 0.0040	616/5	0.0726	0.9968		612/5	-0.0369	0.8387	
31	SNP 4	p52556-rs11057820	Intron 4	GG/AG/AA	G, 0.4871	156/291/172	-0.5049	0.8165		154/290/172	-0.0031	0.8886	
32	SNP 1	p50151-rs2278986	Intron 3	CC/TC/TT	C, 0.2890	56/244/316	0.0131	0.9957	0.7364	56/242/314	0.0020	0.9377	0.8158
32	SNP 2	p50432-chr12_125299088	Intron 3	CC/CT	T, 0.0040	616/5	0.0726	0.9968		612/5	-0.0369	0.8387	
32	SNP 3	p52556-rs11057820	Intron 4	GG/AG/AA	G, 0.4871	156/291/172	-0.5049	0.8165		154/290/172	-0.0031	0.8886	
32	SNP 4	p53093-rs201977189	Exon 5	GA/GG	A, 0.0016	2/615	26.3552	0.3531		2/611	-0.1647	0.5671	
33	SNP 1	p50432-chr12_125299088	Intron 3	CC/CT	T, 0.004	616/5	0.0726	0.9968	0.9134	612/5	-0.0369	0.8387	0.9822
33	SNP 2	p52556-rs11057820	Intron 4	GG/AG/AA	G, 0.4871	156/291/172	-0.5049	0.8165		154/290/172	-0.0031	0.8886	
33	SNP 3	p53093-rs201977189	Exon 5	GA/GG	A, 0.0016	2/615	26.3552	0.3531		2/611	-0.1647	0.5671	
33	SNP 4	p53248-rs140613481	Intron 5	TA/TT	A, 0.0008	1/613	-15.6212	0.6950		1/609	0.6474	0.1093	
34	SNP 1	p52556-rs11057820	Intron 4	GG/AG/AA	G, 0.4871	156/291/172	-0.5049	0.8165	0.6728	154/290/172	-0.0031	0.8886	0.7745
34	SNP 2	p53093-rs201977189	Exon 5	GA/GG	A, 0.0016	2/615	26.3552	0.3531		2/611	-0.1647	0.5671	
34	SNP 3	p53248-rs140613481	Intron 5	TA/TT	A, 0.0008	1/613	-15.6212	0.6950		1/609	0.6474	0.1093	
34	SNP 4	p54492-rs61762481	Intron 5	AA/GA/GG	A, 0.0468	3/52/564	5.4765	0.2940		3/52/560	0.0429	0.4171	
35	SNP 1	p53093-rs201977189	Exon 5	GA/GG	A, 0.0016	2/615	26.3552	0.3531	0.6594	2/611	-0.1647	0.5671	0.7015
35	SNP 2	p53248-rs140613481	Intron 5	TA/TT	A, 0.0008	1/613	-15.6212	0.6950		1/609	0.6474	0.1093	
35	SNP 3	p54492-rs61762481	Intron 5	AA/GA/GG	A, 0.0468	3/52/564	5.4765	0.2940		3/52/560	0.0429	0.4171	
35	SNP 4	p54866-chr12_125294654	Intron 6	GA/GG	A, 0.0008	1/618	-25.4825	0.5241		1/614	-0.2476	0.5419	
36	SNP 1	p53248-rs140613481	Intron 5	TA/TT	A, 0.0008	1/613	-15.6212	0.6950	0.5041	1/609	0.6474	0.1093	0.8108
36	SNP 2	p54492-rs61762481	Intron 5	AA/GA/GG	A, 0.0468	3/52/564	5.4765	0.2940		3/52/560	0.0429	0.4171	
36	SNP 3	p54866-chr12_125294654	Intron 6	GA/GG	A, 0.0008	1/618	-25.4825	0.5241		1/614	-0.2476	0.5419	
36	SNP 4	p55963-rs7134858	Intron 6	TT/CT/CC	T, 0.0759	7/80/532	6.7108	0.1020		7/80/528	-0.0070	0.8663	
37	SNP 1	p54492-rs61762481	Intron 5	AA/GA/GG	A, 0.0468	3/52/564	5.4765	0.2940	0.1956	3/52/560	0.0429	0.4171	0.6943
37	SNP 2	p54866-chr12_125294654	Intron 6	GA/GG	A, 0.0008	1/618	-25.4825	0.5241		1/614	-0.2476	0.5419	
37	SNP 3	p55963-rs7134858	Intron 6	TT/CT/CC	T, 0.0759	7/80/532	6.7108	0.1020		7/80/528	-0.0070	0.8663	
37	SNP 4	p56845-rs838902	Intron 6	AA/AG	G, 0.0211	589/26	-6.0568	0.4492		585/26	0.1231	0.1271	
38	SNP 1	p54866-chr12_125294654	Intron 6	GA/GG	A, 0.0008	1/618	-25.4825	0.5241	0.2082	1/614	-0.2476	0.5419	0.0499
38	SNP 2	p55963-rs7134858	Intron 6	TT/CT/CC	T, 0.0759	7/80/532	6.7108	0.1020		7/80/528	-0.0070	0.8663	
38	SNP 3	p56845-rs838902	Intron 6	AA/AG	G, 0.0211	589/26	-6.0568	0.4492		585/26	0.1231	0.1271	
38	SNP 4	p57308-chr12_125292212	Intron 7	CC/CT	T, 0.0016	615/2	-29.5184	0.2969		611/2	-0.6937	0.0150	
39	SNP 1	p55963-rs7134858	Intron 6	TT/CT/CC	T, 0.0759	7/80/532	6.7108	0.1020	**0.0168**	7/80/528	-0.0070	0.8663	0.3155
39	SNP 2	p56845-rs838902	Intron 6	AA/AG	G, 0.0211	589/26	-6.0568	0.4492		585/26	0.1231	0.1271	
39	SNP 3	p57308-chr12_125292212	Intron 7	CC/CT	T, 0.0016	615/2	-29.5184	0.2969		611/2	-0.6937	0.0150	
39	SNP 4	p57508-rs71458866	Intron 7	GA/GG	A, 0.0162	20/596	5.0155	0.5810		20/592	0.1461	0.1102	
40	SNP 1	p56845-rs838902	Intron 6	AA/AG	G, 0.0211	589/26	-6.0568	0.4492	**0.0178**	585/26	0.1231	0.1271	0.1656
40	SNP 2	p57308-chr12_125292212	Intron 7	CC/CT	T, 0.0016	615/2	-29.5184	0.2969		611/2	-0.6937	0.0150	
40	SNP 3	p57508-rs71458866	Intron 7	GA/GG	A, 0.0162	20/596	5.0155	0.5810		20/592	0.1461	0.1102	
40	SNP 4	p57592-rs838903	Intron 7	GA/GG	A, 0.0161	20/600	5.0154	0.5810		20/596	0.1431	0.1175	
41	SNP 1	p57308-chr12_125292212	Intron 7	CC/CT	T, 0.0016	615/2	-29.5184	0.2969	0.7753	611/2	-0.6937	0.0150	0.1076
41	SNP 2	p57508-rs71458866	Intron 7	GA/GG	A, 0.0162	20/596	5.0155	0.5810		20/592	0.1461	0.1102	
41	SNP 3	p57592-rs838903	Intron 7	GA/GG	A, 0.0161	20/600	5.0154	0.5810		20/596	0.1431	0.1175	
41	SNP 4	p57618-chr12_125291902	Intron 7	GA/GG	A, 0.0008	1/617	27.4039	0.4935		1/613	0.4114	0.3107	
42	SNP 1	p57508-rs71458866	Intron 7	GA/GG	A, 0.0162	20/596	5.0155	0.5810	0.2883	20/592	0.1461	0.1102	0.2674
42	SNP 2	p57592-rs838903	Intron 7	GA/GG	A, 0.0161	20/600	5.0154	0.5810		20/596	0.1431	0.1175	
42	SNP 3	p57618-chr12_125291902	Intron 7	GA/GG	A, 0.0008	1/617	27.4039	0.4935		1/613	0.4114	0.3107	
42	SNP 4	p58514-rs838905	Intron 7	CT/TT	C, 0.0347	43/577	-4.5621	0.4700		43/573	0.0514	0.4190	
43	SNP 1	p57592-rs838903	Intron 7	GA/GG	A, 0.0161	20/600	5.0154	0.5810	0.4824	20/596	0.1431	0.1175	0.4380
43	SNP 2	p57618-chr12_125291902	Intron 7	GA/GG	A, 0.0008	1/617	27.4039	0.4935		1/613	0.4114	0.3107	
43	SNP 3	p58514-rs838905	Intron 7	CT/TT	T, 0.0347	43/577	-4.5621	0.4700		43/573	0.0514	0.4190	
43	SNP 4	p60255-rs3782287	Intron 7	CC/CT/TT	T, 0.4766	176/296/147	0.1466	0.9472		176/294/146	-0.0106	0.6370	
44	SNP 1	p57618-chr12_125291902	Intron 7	GA/GG	A, 0.0008	1/617	27.4039	0.4935	0.6756	1/613	0.4114	0.3107	0.6500
44	SNP 2	p58514-rs838905	Intron 7	CT/TT	T, 0.0347	43/577	-4.5621	0.4700		43/573	0.0514	0.4190	
44	SNP 3	p60255-rs3782287	Intron 7	CC/CT/TT	T, 0.4766	176/296/147	0.1466	0.9472		176/294/146	-0.0106	0.6370	
44	SNP 4	p63483-rs838912	Intron 7	AA/GA/GG	A, 0.4927	156/293/165	0.4413	0.8420		156/291/164	0.0003	0.9905	
45	SNP 1	p58514-rs838905	Intron 7	CT/TT	T, 0.0347	43/577	-4.5621	0.4700	0.9557	43/573	0.0514	0.4190	0.5630
45	SNP 2	p60255-rs3782287	Intron 7	CC/CT/TT	T, 0.4766	176/296/147	0.1466	0.9472		176/294/146	-0.0106	0.6370	
45	SNP 3	p63483-rs838912	Intron 7	AA/GA/GG	A, 0.4927	156/293/165	0.4413	0.8420		156/291/164	0.0003	0.9905	
45	SNP 4	p64285-chr12_125285235	Intron 7	CC/CG	G, 0.0008	615/1	-133.6798	0.0008		611/1	1.0938	0.0065	
46	SNP 1	p60255-rs3782287	Intron 7	CC/CT/TT	T, 0.4766	176/296/147	0.1466	0.9472	0.7495	176/294/146	-0.0106	0.6370	0.6091

46	SNP 2	p63483-rs838912	Intron 7	AA/GA/GG	A, 0.4927	156/293/165	0.4413	0.8420		156/291/164	0.0003	0.9905	
46	SNP 3	p64285-chr12_125285235	Intron 7	CC/CG	G, 0.0008	615/1	-133.6798	0.0008		611/1	1.0938	0.0065	
46	SNP 4	p64772-rs5888	Exon 8	CC/CT/TT	T, 0.4846	169/296/150	0.0119	0.9958		168/293/150	-0.0010	0.9637	
47	SNP 1	p63483-rs838912	Intron 7	AA/GA/GG	A, 0.4927	156/293/165	0.4413	0.8420	0.2973	156/291/164	0.0003	0.9905	0.9886
47	SNP 2	p64285-chr12_125285235	Intron 7	CC/CG	G, 0.0008	615/1	-133.6798	0.0008		611/1	1.0938	0.0065	
47	SNP 3	p64772-rs5888	Exon 8	CC/CT/TT	T, 0.4846	169/296/150	0.0119	0.9958		168/293/150	-0.0010	0.9637	
47	SNP 4	p69612-rs147238482	Intron 8	AG/GG	A, 0.0048	6/617	-8.0419	0.6236		6/613	-0.0667	0.6870	
48	SNP 1	p64285-chr12_125285235	Intron 7	CC/CG	G, 0.0008	615/1	-133.6798	0.0008	0.1831	611/1	1.0938	0.0065	0.5092
48	SNP 2	p64772-rs5888	Exon 8	CC/CT/TT	T, 0.4846	169/296/150	0.0119	0.9958		168/293/150	-0.0010	0.9637	
48	SNP 3	p69612-rs147238482	Intron 8	AG/GG	A, 0.0048	6/617	-8.0419	0.6236		6/613	-0.0667	0.6870	
48	SNP 4	p72551-rs1031605	Intron 9	CC/CT/TT	T, 0.1755	421/182/18	0.9119	0.7593		419/181/18	-0.0518	0.0885	
49	SNP 1	p64772-rs5888	Exon 8	CC/CT/TT	T, 0.4846	169/296/150	0.0119	0.9958	0.2563	168/293/150	-0.0010	0.9637	0.5902
49	SNP 2	p69612-rs147238482	Intron 8	AG/GG	A, 0.0048	6/617	-8.0419	0.6236		6/613	-0.0667	0.6870	
49	SNP 3	p72551-rs1031605	Intron 9	CC/CT/TT	T, 0.1755	421/182/18	0.9119	0.7593		419/181/18	-0.0518	0.0885	
49	SNP 4	p76757-rs9919713	Intron 9	AA/AT/TT	T, 0.0379	575/43/2	-2.4875	0.6663		571/43/2	0.0439	0.4535	
50	SNP 1	p69612-rs147238482	Intron 8	AG/GG	A, 0.0048	6/617	-8.0419	0.6236	0.8969	6/613	-0.0667	0.6870	0.3894
50	SNP 2	p72551-rs1031605	Intron 9	CC/CT/TT	T, 0.1755	421/182/18	0.9119	0.7593		419/181/18	-0.0518	0.0885	
50	SNP 3	p76757-rs9919713	Intron 9	AA/AT/TT	T, 0.0379	575/43/2	-2.4875	0.6663		571/43/2	0.0439	0.4535	
50	SNP 4	p77250-rs201901986	Intron 9	GG/GT	T, 0.0225	594/28	-6.9460	0.3696		590/28	0.0796	0.3076	
51	SNP 1	p72551-rs1031605	Intron 9	CC/CT/TT	T, 0.1755	421/182/18	0.9119	0.7593	0.0682	419/181/18	-0.0518	0.0885	0.4344
51	SNP 2	p76757-rs9919713	Intron 9	AA/AT/TT	T, 0.0379	575/43/2	-2.4875	0.6663		571/43/2	0.0439	0.4535	
51	SNP 3	p77250-rs201901986	Intron 9	GG/GT	T, 0.0225	594/28	-6.9460	0.3696		590/28	0.0796	0.3076	
51	SNP 4	p77251-rs34339961	Intron 9	AA/AT	T, 0.0226	592/28	-7.0407	0.3632		588/28	0.0774	0.3210	
52	SNP 1	p76757-rs9919713	Intron 9	AA/AT/TT	T, 0.0379	575/43/2	-2.4875	0.6663	0.6202	571/43/2	0.0439	0.4535	0.5989
52	SNP 2	p77250-rs201901986	Intron 9	GG/GT	T, 0.0225	594/28	-6.9460	0.3696		590/28	0.0796	0.3076	
52	SNP 3	p77251-rs34339961	Intron 9	AA/AT	T, 0.0226	592/28	-7.0407	0.3632		588/28	0.0774	0.3210	
52	SNP 4	p77842-rs2272310	Intron 10	AA/GA/GG	A, 0.0146	1/16/599	3.3863	0.7064		1/16/595	0.0100	0.9132	
53	SNP 1	p77250-rs201901986	Intron 9	GG/GT	T, 0.0225	594/28	-6.9460	0.3696	0.4260	590/28	0.0796	0.3076	0.8024
53	SNP 2	p77251-rs34339961	Intron 9	AA/AT	T, 0.0226	592/28	-7.0407	0.3632		588/28	0.0774	0.3210	
53	SNP 3	p77842-rs2272310	Intron 10	AA/GA/GG	A, 0.0146	1/16/599	3.3863	0.7064		1/16/595	0.0100	0.9132	
53	SNP 4	p78334-chr12_125271186	Intron 10	TG/TT	G, 0.0024	3/620	28.0847	0.2247		3/616	0.2019	0.3879	
54	SNP 1	p77251-rs34339961	Intron 9	AA/AT	T, 0.0226	592/28	-7.0407	0.3632	0.2576	588/28	0.0774	0.3210	0.7195
54	SNP 2	p77842-rs2272310	Intron 10	AA/GA/GG	A, 0.0146	1/16/599	3.3863	0.7064		1/16/595	0.0100	0.9132	
54	SNP 3	p78334-chr12_125271186	Intron 10	TG/TT	G, 0.0024	3/620	28.0847	0.2247		3/616	0.2019	0.3879	
54	SNP 4	p78430-rs838897	Intron 10	GG/CG/CC	G, 0.0364	2/41/575	-2.0316	0.7298		2/41/571	0.0439	0.4626	
55	SNP 1	p77842-rs2272310	Intron 10	AA/GA/GG	A, 0.0146	1/16/599	3.3863	0.7064	0.4277	1/16/595	0.0100	0.9132	0.9328
55	SNP 2	p78334-chr12_125271186	Intron 10	TG/TT	G, 0.0024	3/620	28.0847	0.2247		3/616	0.2019	0.3879	
55	SNP 3	p78430-rs838897	Intron 10	GG/CG/CC	G, 0.0364	2/41/575	-2.0316	0.7298		2/41/571	0.0439	0.4626	
55	SNP 4	p78747-rs2293440	Intron 11	CC/TC/TT	C, 0.0243	1/28/589	-5.4568	0.4507		1/28/585	0.0415	0.5709	
56	SNP 1	p78334-chr12_125271186	Intron 10	TG/TT	G, 0.0024	3/620	28.0847	0.2247	0.5589	3/616	0.2019	0.3879	0.6392
56	SNP 2	p78430-rs838897	Intron 10	GG/CG/CC	G, 0.0364	2/41/575	-2.0316	0.7298		2/41/571	0.0439	0.4626	
56	SNP 3	p78747-rs2293440	Intron 11	CC/TC/TT	C, 0.0243	1/28/589	-5.4568	0.4507		1/28/585	0.0415	0.5709	
56	SNP 4	p79721-rs838896	Intron 11	GG/GC/CC	C, 0.3169	293/257/67	2.7275	0.2564		292/254/67	0.0201	0.4080	
57	SNP 1	p78430-rs838897	Intron 10	GG/CG/CC	G, 0.0364	2/41/575	-2.0316	0.7298	0.8618	2/41/571	0.0439	0.4626	0.5842
57	SNP 2	p78747-rs2293440	Intron 11	CC/TC/TT	C, 0.0243	1/28/589	-5.4568	0.4507		1/28/585	0.0415	0.5709	
57	SNP 3	p79721-rs838896	Intron 11	GG/GC/CC	C, 0.3169	293/257/67	2.7275	0.2564		292/254/67	0.0201	0.4080	
57	SNP 4	p79828-rs838895	Intron 11	GG/CG/CC	G, 0.2891	54/243/310	3.5525	0.1489		54/242/309	0.0060	0.8131	
58	SNP 1	p78747-rs2293440	Intron 11	CC/TC/TT	C, 0.0243	1/28/589	-5.4568	0.4507	0.8585	1/28/585	0.0415	0.5709	0.6697
58	SNP 2	p79721-rs838896	Intron 11	GG/GC/CC	C, 0.3169	293/257/67	2.7275	0.2564		292/254/67	0.0201	0.4080	
58	SNP 3	p79828-rs838895	Intron 11	GG/CG/CC	G, 0.2891	54/243/310	3.5525	0.1489		54/242/309	0.0060	0.8131	
58	SNP 4	p80045-rs838893	Intron 11	GG/GA/AA	A, 0.2879	316/251/53	2.4968	0.3131		315/248/53	0.0091	0.7174	
59	SNP 1	p79721-rs838896	Intron 11	GG/GC/CC	C, 0.3169	293/257/67	2.7275	0.2564	0.4501	292/254/67	0.0201	0.4080	0.5004
59	SNP 2	p79828-rs838895	Intron 11	GG/CG/CC	G, 0.2891	54/243/310	3.5525	0.1489		54/242/309	0.0060	0.8131	
59	SNP 3	p80045-rs838893	Intron 11	GG/GA/AA	A, 0.2879	316/251/53	2.4968	0.3131		315/248/53	0.0091	0.7174	
59	SNP 4	p83088-rs7977729	Intron 12	GG/AG/AA	G, 0.2669	44/243/333	3.0674	0.2329		44/240/332	0.0042	0.8720	
60	SNP 1	p79828-rs838895	Intron 11	GG/CG/CC	G, 0.2891	54/243/310	3.5525	0.1489	0.4922	54/242/309	0.0060	0.8131	0.2926
60	SNP 2	p80045-rs838893	Intron 11	GG/GA/AA	A, 0.2879	316/251/53	2.4968	0.3131		315/248/53	0.0091	0.7174	
60	SNP 3	p83088-rs7977729	Intron 12	GG/AG/AA	G, 0.2669	44/243/333	3.0674	0.2329		44/240/332	0.0042	0.8720	
60	SNP 4	p83884-rs701106	Intron 12	TT/CT/CC	T, 0.1527	17/156/449	-1.4985	0.6266		17/155/446	-0.0407	0.1948	
61	SNP 1	p80045-rs838893	Intron 11	GG/GA/AA	A, 0.2879	316/251/53	2.4968	0.3131	0.8514	315/248/53	0.0091	0.7174	0.2215
61	SNP 2	p83088-rs7977729	Intron 12	GG/AG/AA	G, 0.2669	44/243/333	3.0674	0.2329		44/240/332	0.0042	0.8720	
61	SNP 3	p83884-rs701106	Intron 12	TT/CT/CC	T, 0.1527	17/156/449	-1.4985	0.6266		17/155/446	-0.0407	0.1948	
61	SNP 4	p86436-rs10396214	Exon 13-3' UTR	CC/CT	T, 0.0088	612/11	0.5238	0.9658		609/10	-0.0630	0.6251	

62	SNP 1	p83088-rs7977729	Intron 12	GG/AG/AA	G, 0.2669	44/243/333	3.0674	0.2329	0.7858	44/240/332	0.0042	0.8720	0.6087
62	SNP 2	p83884-rs701106	Intron 12	TT/CT/CC	T, 0.1527	17/156/449	-1.4985	0.6266		17/155/446	-0.0407	0.1948	
62	SNP 3	p86436-rs10396214	Exon 13-3' UTR	CC/CT	T, 0.0088	612/11	0.5238	0.9658		609/10	-0.0630	0.6251	
62	SNP 4	p87004-rs184715678	Exon 13-3' UTR	CA/CC	A, 0.0016	2/619	17.6266	0.5332		2/615	0.2703	0.3432	
63	SNP 1	p83884-rs701106	Intron 12	TT/CT/CC	T, 0.1527	17/156/449	-1.4985	0.6266	0.8760	17/155/446	-0.0407	0.1948	0.4783
63	SNP 2	p86436-rs10396214	Exon 13-3' UTR	CC/CT	T, 0.0088	612/11	0.5238	0.9658		609/10	-0.0630	0.6251	
63	SNP 3	p87004-rs184715678	Exon 13-3' UTR	CA/CC	A, 0.0016	2/619	17.6266	0.5332		2/615	0.2703	0.3432	
63	SNP 4	p87210-chr12_125262310	Exon 13-3' UTR	GG/GA	A, 0.0016	621/2	-15.0364	0.5960		617/2	0.0583	0.8389	
64	SNP 1	p86436-rs10396214	Exon 13-3' UTR	CC/CT	T, 0.0088	612/11	0.5238	0.9658	0.9506	609/10	-0.0630	0.6251	0.9027
64	SNP 2	p87004-rs184715678	Exon 13-3' UTR	CA/CC	A, 0.0016	2/619	17.6266	0.5332		2/615	0.2703	0.3432	
64	SNP 3	p87210-chr12_125262310	Exon 13-3' UTR	GG/GA	A, 0.0016	621/2	-15.0364	0.5960		617/2	0.0583	0.8389	
64	SNP 4	p87381-rs192190977	3' flanking	CC/CT	T, 0.0024	618/3	6.9443	0.7645		614/3	-0.2773	0.2352	
65	SNP 1	p87004-rs184715678	Exon 13-3' UTR	CA/CC	A, 0.0016	2/619	17.6266	0.5332	0.9649	2/615	0.2703	0.3432	0.6197
65	SNP 2	p87210-chr12_125262310	Exon 13-3' UTR	GG/GA	A, 0.0016	621/2	-15.0364	0.5960		617/2	0.0583	0.8389	
65	SNP 3	p87381-rs192190977	3' flanking	CC/CT	T, 0.0024	618/3	6.9443	0.7645		614/3	-0.2773	0.2352	
65	SNP 4	p87681-rs838883	3' flanking	GG/GA/AA	A, 0.0845	518/101/2	-0.4540	0.9132		514/101/2	-0.0403	0.3400	
66	SNP 1	p87210-chr12_125262310	Exon 13-3' UTR	GG/GA	A, 0.0016	621/2	-15.0364	0.5960	0.8725	617/2	0.0583	0.8389	0.6312
66	SNP 2	p87381-rs192190977	3' flanking	CC/CT	T, 0.0024	618/3	6.9443	0.7645		614/3	-0.2773	0.2352	
66	SNP 3	p87681-rs838883	3' flanking	GG/GA/AA	A, 0.0845	518/101/2	-0.4540	0.9132		514/101/2	-0.0403	0.3400	
66	SNP 4	p87927-rs838880	3' flanking	AA/AG/GG	G, 0.3237	281/278/62	-1.9523	0.4256		279/276/62	-0.0197	0.4247	

GT; genotype; LDL-C, low-density lipoprotein cholesterol; MA, minor allele; MAF, minor allele frequency; SNP, single nucleotide polymorphism; TG, triglycerides; UTR, untranslated region.
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. TG values were Box-Cox transformed. Results were adjusted for covariates (LDL-C: age and body mass index; TG: sex, age, smoking, and body mass index).
Single-site *P*-values were unadjusted for multiple testing, see details in Tables A10 and A11.
Significant global *P*-values (<0.05) are shown in **bold**.
*Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
†dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).

Table A16. Summary of RegulomeDB scores of 80 *SCARB1* identified variants (44 sequence variants, 32 common HapMap-CEU tagSNPs, and 4 additional relevant variants from the literatures).

RegulomeDB Score*	No of Variants						Total N (%)
	MAF ≥5% (n = 45)		MAF between 1-5% (n = 13)		MAF ≤1% (n = 22)		
	Coding	Non-coding	Coding	Non-coding	Coding	Non-coding	
1f		1					1 (1.25)
2a							
2b		1				4	5 (6.25)
3a	1					1	2 (2.50)
3b		1					1 (1.25)
4	1	5		5	1	7	19 (23.75)
5		22		5	3	2	32 (40.00)
6		7		3		1	11 (13.75)
7		6				3	9 (11.25)
Total N	**2**	**43**		**13**	**4**	**18**	**80 (100.00)**

CEU, Utah residents with northern and western European ancestry; MAF, minor allele frequency; SNP, single nucleotide polymorphism.
A list of 44 sequence variants is shown in Table A5.
A list of 32 common HapMap-CEU tagSNPs is shown in Table A4.
*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 A17. RegulomeDB scores and functional assignments of 80 *SCARB1* identified variants (44 sequence variants, 32 common HapMap-CEU tagSNPs, and 4 additional relevant variants from the literatures).

SNP Name*	SNP ID†	Chr12 Position*	Location	Amino Acid Change	Major/Minor Alleles	MAF	Associated Trait‡	RegulomeDB Score§	RegulomeDB Functional Assignment
p1257	rs4238001	125348263	Exon 1	Gly2Ser	G/A	0.0818		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|TFAP2C, Protein_Binding|ChIP-seq|POU2F2, Protein_Binding|ChIP-seq|MXI1, Protein_Binding|ChIP-seq|CDX2, Protein_Binding|ChIP-seq|GATA1, Protein_Binding|ChIP-seq|TFAP2A, Protein_Binding|ChIP-seq|RAD21, Protein_Binding|ChIP-seq|E2F6, Protein_Binding|ChIP-seq|TAF1, Protein_Binding|ChIP-seq|CCNT2, Protein_Binding|ChIP-seq|POLR2A
p1588		125347932	Intron 1		G/C	0.0008		4	Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|MAX, Protein_Binding|ChIP-seq|E2F1
p4072	rs7139401	125345448	Intron 1		C/T	0.4564		3b	Motifs|PWM|Mafk, Motifs|PWM|Dlx3, Motifs|PWM|Dlx4, Protein_Binding|ChIP-seq|MAFK
p5055	rs11057869	125344465	Intron 1		G/A	0.0875		7	No data
p7650	rs11615630	125341870	Intron 1		G/A	0.3817		5	Chromatin_Structure|DNase-seq
p10292	rs4765181	125339228	Intron 1		G/T	0.3972		5	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq
p13570	rs11057864	125335950	Intron 1		G/T	0.0427		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|HNF4A
p16565	rs10773111	125332955	Intron 1		G/A	0.4249		6	Single_Nucleotides|SCARB1|eQTL
p20207	rs11057853	125329313	Intron 1		A/G	0.4269		5	Motifs|PWM|HNF4, Motifs|PWM|HNF4A, Motifs|PWM|HNF4directrepeat1, Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq
p20741	rs11057851	125328779	Intron 1		C/T	0.1157		5	Chromatin_Structure|DNase-seq
p21145	rs3924313	125328375	Intron 1		C/T	0.3083		6	Motifs|PWM|TFIIA
p22116	rs12370382	125327404	Intron 1		G/A	0.4005		1f	Single_Nucleotides|SCARB1|eQTL, Chromatin_Structure|DNase-seq

p26525	rs10773109	125322995	Intron 1		C/G	0.4176		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|JUN, Protein_Binding|ChIP-seq|TFAP2C, Protein_Binding|ChIP-seq|CEBPB, Protein_Binding|ChIP-seq|STAT1, Protein_Binding|ChIP-seq|TBP, Protein_Binding|ChIP-seq|FOXA1, Protein_Binding|ChIP-seq|GATA3, Protein_Binding|ChIP-seq|MXI1, Protein_Binding|ChIP-seq|EP300, Protein_Binding|ChIP-seq|MAX, Protein_Binding|ChIP-seq|USF1, Protein_Binding|ChIP-seq|ESR1, Protein_Binding|ChIP-seq|STAT3, Protein_Binding|ChIP-seq|HNF4A, Protein_Binding|ChIP-seq|TFAP2A, Protein_Binding|ChIP-seq|MYC, Protein_Binding|ChIP-seq|TRIM28, Protein_Binding|ChIP-seq|USF2, Protein_Binding|ChIP-seq|TCF4, Protein_Binding|ChIP-seq|RFX5, Protein_Binding|ChIP-seq|NR3C1, Protein_Binding|ChIP-seq|FOSL2, Protein_Binding|ChIP-seq|RXRA, Protein_Binding|ChIP-seq|HDAC2, Protein_Binding|ChIP-seq|ZNF263, Protein_Binding|ChIP-seq|SP1, Protein_Binding|ChIP-seq|JUND, Protein_Binding|ChIP-seq|GTF2F1, Protein_Binding|ChIP-seq|POLR2A
p28137	rs12229555	125321383	Intron 1		A/G	0.2188		7	No data
p28692	rs4765622	125320828	Intron 1		C/T	0.4693		5	Chromatin_Structure|DNase-seq
p28957	rs11057844	125320563	Intron 1		G/A	0.1839	HDL-C	5	Motifs|Footprinting|HTF, Motifs|PWM|HTF, Protein_Binding|ChIP-seq|GATA1
p31072	rs10846749	125318448	Intron 1		C/G	0.3448		4	Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|NANOG, Protein_Binding|ChIP-seq|EP300, Protein_Binding|ChIP-seq|MAX, Protein_Binding|ChIP-seq|USF1, Protein_Binding|ChIP-seq|TCF12, Protein_Binding|ChIP-seq|TCF4, Protein_Binding|ChIP-seq|RXRA, Protein_Binding|ChIP-seq|SP1, Protein_Binding|ChIP-seq|JUND
p31938	rs10744182	125317582	Intron 1		A/G	0.4130		5	Chromatin_Structure|DNase-seq
p32129	rs10773107	125317391	Intron 1		G/T	0.4828		7	No data
p32273	rs12580803	125317247	Intron 1		T/C	0.1726		5	Chromatin_Structure|DNase-seq
p32395	rs12581963	125317125	Intron 1		C/T	0.0829		5	Motifs|PWM|E2F-1, Chromatin_Structure|DNase-seq
p32860	rs7967406	125316660	Intron 1		A/C	0.3921		6	Motifs|PWM|Hbp1, Motifs|PWM|Rfx3
p36908	rs10846745	125312612	Intron 1		C/G	0.4748		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|TFAP2C, Protein_Binding|ChIP-seq|AR, Protein_Binding|ChIP-seq|FOXA1, Protein_Binding|ChIP-seq|EP300, Protein_Binding|ChIP-seq|CDX2, Protein_Binding|ChIP-seq|HNF4A
p37095	rs10846744	125312425	Intron 1		C/G	0.1364		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|TFAP2C, Protein_Binding|ChIP-seq|AR, Protein_Binding|ChIP-seq|FOXA1, Protein_Binding|ChIP-seq|GATA3, Protein_Binding|ChIP-seq|EP300, Protein_Binding|ChIP-seq|MAX, Protein_Binding|ChIP-seq|ESR1, Protein_Binding|ChIP-seq|CDX2, Protein_Binding|ChIP-seq|HNF4A
p42467	rs11057830	125307053	Intron 1		C/T	0.1329		7	No data
p47235	rs199779577	125302285	Intron 1		C/T	0.0016		4	Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|IKZF1
p47416	rs73227571	125302104	Exon 2	Tyr92Tyr	C/T	0.0008		5	Chromatin_Structure|DNase-seq

p48969	rs2343394	125300551	Intron 2		C/T	0.2850	ApoB	5	Chromatin_Structure|DNase-seq
p49518	rs144194221	125300002	Intron 2		G/A	0.0217		5	Motifs|Footprinting|MTF-1, Motifs|PWM|MTF-1, Chromatin_Structure|DNase-seq
p49690	rs4765615	125299830	Intron 2		A/G	0.4497	ApoB	5	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq
p49978	rs5891	125299542	Exon 3	Val135Ile	G/A	0.0096		5	Motifs|PWM|Pax-3, Motifs|Footprinting|Pax-3, Chromatin_Structure|DNase-seq
p50151	rs2278986	125299369	Intron 3		T/C	0.2890	ApoB	5	Chromatin_Structure|DNase-seq
p50432		125299088	Intron 3		C/T	0.0040		2b	Motifs|Footprinting|Myc, Motifs|PWM|c-Myc:Max, Motifs|PWM|N-Myc, Motifs|Footprinting|Mycn, Motifs|PWM|Mycn, Motifs|PWM|Ebox, Motifs|Footprinting|CLOCK:BMAL, Motifs|PWM|USF, Motifs|PWM|Myc, Motifs|PWM|CLOCK:BMAL, Motifs|Footprinting|N-Myc, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|IKZF1
p52556	rs11057820	125296964	Intron 4		A/G	0.4871	ApoB	5	Chromatin_Structure|DNase-seq
p53093	rs201977189	125296427	Exon 5	Gly239Arg	G/A	0.0016		5	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq
p53158[ll]	rs61932577	125296362	Intron 5		C/T	0.091		5	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq
p53248	rs140613481	125296272	Intron 5		T/A	0.0008		5	Chromatin_Structure|DNase-seq
p53440[ll]	rs55834271	125296080	Intron 5		T/C	0.269		6	Motifs|PWM|Lyf-1, Motifs|PWM|Ikaros, Motifs|Footprinting|Ikaros, Motifs|Footprinting|Lyf-1
p53490[ll]		125296030	Intron 5		C/A	0.005		7	No data
p54492	rs61762481	125295028	Intron 5		G/A	0.0468		4	Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|RFX3
p54866		125294654	Intron 6		G/A	0.0008		4	Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|RFX3, Protein_Binding|ChIP-seq|STAT3
p55963	rs7134858	125293557	Intron 6		C/T	0.0759		6	Motifs|PWM|STAT1
p56845	rs838902	125292675	Intron 6		A/G	0.0211		5	Motifs|PWM|Bbx, Chromatin_Structure|DNase-seq
p57308		125292212	Intron 7		C/T	0.0016		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|GATA1, Protein_Binding|ChIP-seq|NR3C1, Protein_Binding|ChIP-seq|TAL1, Protein_Binding|ChIP-seq|GATA6, Protein_Binding|ChIP-seq|CCNT2, Protein_Binding|ChIP-seq|GATA2
p57508	rs71458866	125292012	Intron 7		G/A	0.0162		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|GATA1
p57592	rs838903	125291928	Intron 7		G/A	0.0161		4	Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|GATA1
p57618		125291902	Intron 7		G/A	0.0008		4	Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|GATA1
p58514	rs838905	125291006	Intron 7		T/C	0.0347		4	Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|CEBPB
p60255	rs3782287	125289265	Intron 7		C/T	0.4766		5	Chromatin_Structure|DNase-seq
p63483	rs838912	125286037	Intron 7		G/A	0.4927		7	No data
p64285		125285235	Intron 7		C/G	0.0008		3a	Motifs|PWM|CTCF, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|RFX3
p64772	rs5888	125284748	Exon 8	Ala350Ala	C/T	0.4846		3a	Motifs|PWM|Nr2f2, Motifs|PWM|PPARalpha:RXRalpha, Motifs|PWM|Rara, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|RFX3, Protein_Binding|ChIP-seq|GATA2
p69612	rs147238482	125279908	Intron 8		G/A	0.0048		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|EBF1, Protein_Binding|ChIP-seq|NFKB1, Protein_Binding|ChIP-seq|TCF12

p70038 del5[‖] [70038_ 70042]		125279482	Intron 9		[GTTTT/-]	0.005		6	Motifs|PWM|Foxl1, Motifs|PWM|RREB1, Motifs|PWM|FOXO1, Motifs|PWM|Foxj3, Motifs|PWM|Nanog, Motifs|PWM|FOXP1, Motifs|Footprinting|FOXO1
p70088[‖]	rs117585141	125279432	Intron 9		G/A	0.005		7	No data
p70095[‖]		125279425	Intron 9		T/C	0.005		7	No data
p70292[‖]	rs150388176	125279228	Intron 9		C/T	0.005		5	Chromatin_Structure|DNase-seq
p72551	rs1031605	125276969	Intron 9		C/T	0.1755		5	Motifs|PWM|HIC1, Chromatin_Structure|DNase-seq
p76757	rs9919713	125272763	Intron 9		A/T	0.0379		6	Motifs|PWM|Alx-4, Motifs|PWM|FOXJ2, Motifs|Footprinting|FOXJ2, Motifs|Footprinting|Alx-4
p77250	rs201901986	125272270	Intron 9		G/T	0.0225		6	Motifs|PWM|Mtf1, Motifs|PWM|Tcfap2e, Motifs|PWM|Srf
p77251	rs34339961	125272269	Intron 9		A/T	0.0226		6	Motifs|PWM|Mtf1, Motifs|PWM|Tcfap2e, Motifs|PWM|Srf
p77842	rs2272310	125271678	Intron 10		G/A	0.0146		5	Chromatin_Structure|DNase-seq
p78334		125271186	Intron 10		T/G	0.0024		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|TBP, Protein_Binding|ChIP-seq|USF1, Protein_Binding|ChIP-seq|USF2, Protein_Binding|ChIP-seq|POLR2A
p78430	rs838897	125271090	Intron 10		C/G	0.0364		5	Motifs|PWM|AP-2, Motifs|Footprinting|AP-2, Motifs|PWM|RNF96, Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq
p78747	rs2293440	125270773	Intron 11		T/C	0.0243		5	Chromatin_Structure|DNase-seq
p79721	rs838896	125269799	Intron 11		G/C	0.3169		5	Chromatin_Structure|DNase-seq
p79828	rs838895	125269692	Intron 11		C/G	0.2891		5	Motifs|PWM|Pax-5, Chromatin_Structure|DNase-seq
p80045	rs838893	125269475	Intron 11		G/A	0.2879		5	Chromatin_Structure|DNase-seq
p83088	rs7977729	125266432	Intron 12		A/G	0.2669		6	Motifs|Footprinting|HFH3(FOXI1), Motifs|Footprinting|HFH4(FOXJ1), Motifs|Footprinting|POU3F2, Motifs|Footprinting|HNF3, Motifs|PWM|HFH4(FOXJ1), Motifs|PWM|HFH3(FOXI1), Motifs|PWM|HNF3, Motifs|PWM|FOXJ2, Motifs|Footprinting|Cdx, Motifs|PWM|Cdx, Motifs|PWM|POU3F2
p83884	rs701106	125265636	Intron 12		C/T	0.1527	HDL-C	5	Chromatin_Structure|DNase-seq
p86316[¶]	rs701104	125263204	Intron 12		G/T	0.105		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|EGR1, Protein_Binding|ChIP-seq|BATF
p86436	rs10396214	125263084	Exon 13-3' UTR	Arg484Trp[#]	C/T	0.0088		4	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|EGR1
p87004	rs184715678	125262516	Exon 13-3' UTR		C/A	0.0016		2b	Motifs|Footprinting|SP1, Motifs|PWM|SP1, Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|HEY1, Protein_Binding|ChIP-seq|HNF4A, Protein_Binding|ChIP-seq|TCF4, Protein_Binding|ChIP-seq|FOSL2, Protein_Binding|ChIP-seq|HDAC2, Protein_Binding|ChIP-seq|JUND, Protein_Binding|ChIP-seq|POLR2A, Protein_Binding|ChIP-seq|HNF4G
p87210		125262310	Exon 13-3' UTR		G/A	0.0016		2b	Motifs|PWM|Zfx, Motifs|PWM|AP-2, Motifs|Footprinting|AP-2, Motifs|Footprinting|Zfx, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|TCF4

p87381	rs192190977	125262139	3' flanking	C/T	0.0024		2b	Motifs|PWM|Zfp740, Motifs|PWM|PPARalpha:RXRalpha, Motifs|Footprinting|UF1H3BETA, Motifs|Footprinting|PPARalpha:RXRalpha, Motifs|PWM|Zfp281, Motifs|PWM|UF1H3BETA, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|FOXA2, Protein_Binding|ChIP-seq|FOXA1, Protein_Binding|ChIP-seq|ELF1
p87416¶	rs838884	125262104	3' flanking	T/C	0.335		2b	Motifs|PWM|DEAF1, Motifs|Footprinting|DEAF1, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|CEBPB, Protein_Binding|ChIP-seq|FOXA2, Protein_Binding|ChIP-seq|FOXA1, Protein_Binding|ChIP-seq|ELF1, Protein_Binding|ChIP-seq|FOSL2, Protein_Binding|ChIP-seq|JUND
p87681	rs838883	125261839	3' flanking	G/A	0.0845		5	Chromatin_Structure|FAIRE, Chromatin_Structure|DNase-seq
p87707¶	rs838882	125261813	3' flanking	A/G	0.328		7	No data
p87723¶	rs838881	125261797	3' flanking	T/C	0.330		6	Motifs|PWM|NFIL3, Motifs|Footprinting|NFIL3, Motifs|PWM|TFIIA, Chromatin_Structure|FAIRE
p87927	rs838880	125261593	3' flanking	A/G	0.3237	HDL-C	5	Chromatin_Structure|DNase-seq

ApoB, apolipoprotein B; CEU, Utah residents with northern and western European ancestry; del, deletion; eQTL, expression quantitative trait loci; HDL-C, high-density lipoprotein cholesterol; MAF, minor allele frequency; SNP, single nucleotide polymorphism; UTR, untranslated region.
A list of 44 sequence variants is shown in Table A5.
A list of 32 common HapMap-CEU tagSNPs is shown in Table A4.
Underlined MAFs were derived from sequencing data due to not chosen for genotyping or genotyping failure; see details of selected variants in Tables A4 and A5.
*Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
†dbSNP build 139: GRCh37.p10; 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA).
‡SNPs with MAF ≥5% that yielded a significance threshold of single-site association (P <0.05; Tables A8-A11) with any of the 4 lipid traits in this study.
§Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table A16 or can be seen at http://regulome.stanford.edu/help.
||The variants (n = 7) were identified by sequencing, but failed genotyping.
¶Sequence variants (n = 4) that not chosen for genotyping based on linkage disequilibrium structure (Figure A2).
#rs10396214 is translated leading to amino acid change (Arg484Trp) in isoform 2 (alternative splice form); not translated in isoform 1.

Table A18. RegulomeDB scores and functional assignments of *SCARB1* common HapMap-CEU* tagSNPs with significant association for HDL-C and ApoB and their alleles captured.

Bin #	Type of SNPs	SNP ID*/ Associated Trait†	Chr12 Position‡	RegulomeDB Score§	RegulomeDB Functional Assignment
4	TagSNP	**rs838880**/ HDL-C	125261593	5	Chromatin_Structure|DNase-seq
	SNP captured	rs838884	125262104	2b	Motifs|PWM|DEAF1, Motifs|Footprinting|DEAF1, Chromatin_Structure|DNase-seq, Protein_Binding|ChIP-seq|JUND, Protein_Binding|ChIP-seq|CEBPB, Protein_Binding|ChIP-seq|FOSL2, Protein_Binding|ChIP-seq|FOXA1, Protein_Binding|ChIP-seq|ELF1, Protein_Binding|ChIP-seq|FOXA2
	SNP captured	rs838882	125261813	7	No data
	SNP captured	rs838881	125261797	6	Motifs|PWM|TFIIA, Motifs|PWM|NFIL3, Motifs|Footprinting|NFIL3, Chromatin_Structure|FAIRE
	SNP captured	rs838879	125261441	7	No data
12	TagSNP	**rs2343394**/ ApoB	125300551	5	Chromatin_Structure|DNase-seq
	SNP captured	rs745529	125300472	5	Chromatin_Structure|DNase-seq
13	TagSNP	**rs11057844**/ HDL-C	125320563	5	Motifs|Footprinting|HTF, Motifs|PWM|HTF, Protein_Binding|ChIP-seq|GATA1
	SNP captured	rs7954519	125320155	7	No data

ApoB, apolipoprotein B; CEU, Utah residents with northern and western European ancestry; HDL-C, high-density lipoprotein cholesterol; SNP, single nucleotide polymorphism.
Bins of common HapMap-CEU tagSNPs and SNPs captured were based on the result of Haploview Tagger analysis, see details in Table A4 and Figure A1.
HapMap-CEU tagSNPs with significant evidence of single-site association (*P* <0.05; Table 6.2 and Tables A4, A8-A11) observed in this study are shown in **bold**.
*HapMap-CEU release #27, genome build 36, dbSNP build 126.
†Based on HapMap-CEU tagSNPs with significant evidence of single-site association (*P* <0.05; Tables A4, A8-A11) with any of the 4 lipid traits in this study.
‡Based on the *SCARB1* RefSeq (hg19, NM_005505) from CHIP Bioinformatics.
§Detailed RegulomeDB (version 1.0) scoring scheme is described in the footnote of Table A16 or can be seen at http://regulome.stanford.edu/help.

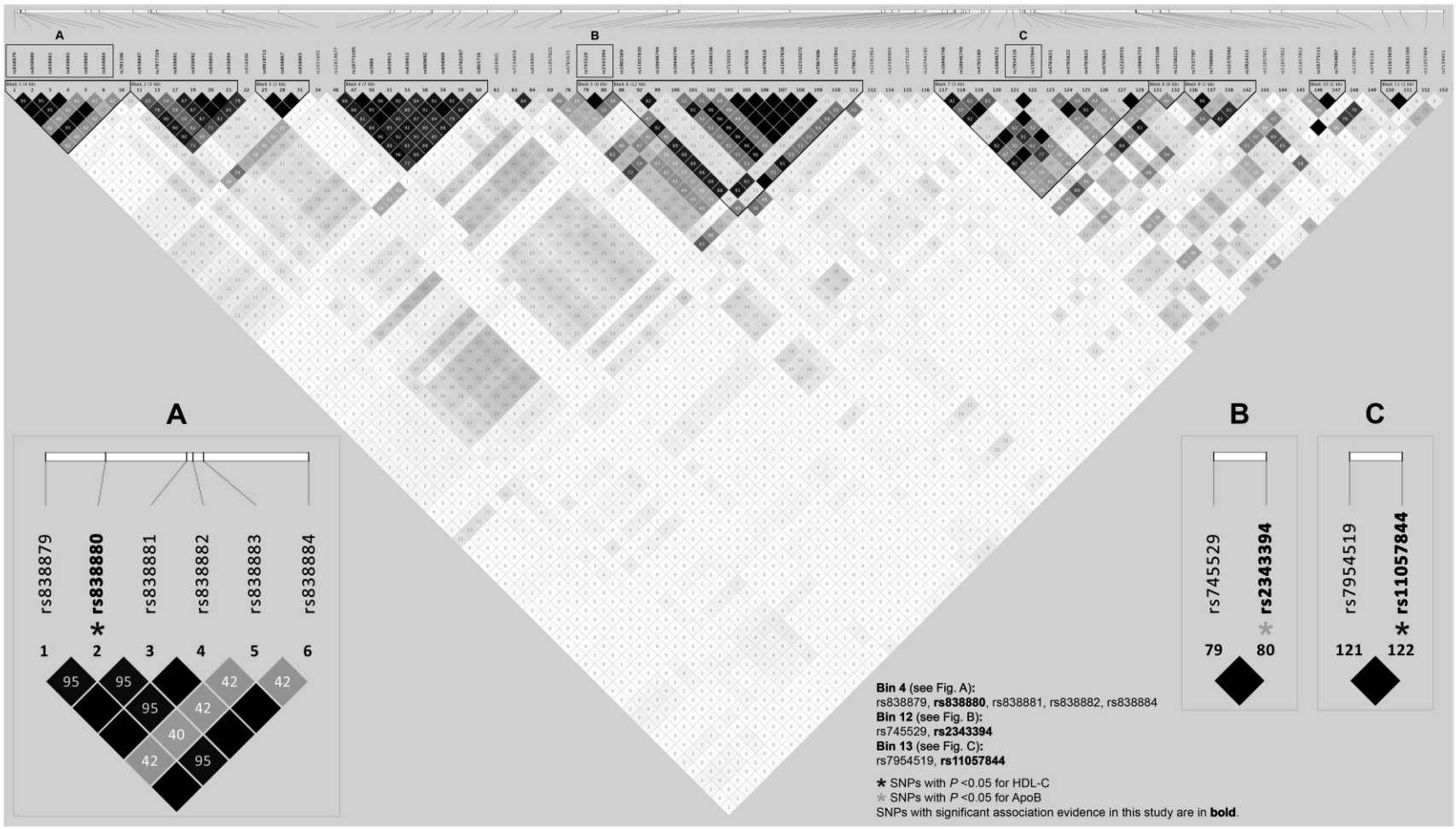

A
B
C
A
rs838879
rs838880
rs838881
rs838882
rs838883
rs838884
1
2
3
4
5
6
95
95
95
42
42
42
40
42
95
B
rs745529
rs2343394
79
80
C
rs7954519
rs11057844
121
122
Bin 4 (see Fig. A):
rs838879, rs838880, rs838881, rs838882, rs838884
Bin 12 (see Fig. B):
rs745529, rs2343394
Bin 13 (see Fig. C):
rs7954519, rs11057844
* SNPs with P <0.05 for HDL-C
* SNPs with P <0.05 for ApoB
SNPs with significant association evidence in this study are in bold.

Figure A1. Linkage disequilibrium structure of 81 *SCARB1* common HapMap-CEU tagSNPs (HapMap release #27, genome build 36, dbSNP build 126).

Using Haploview Tagger analysis with MAF ≥5% and r^2 ≥0.80, 35 bins were obtained from a total of 81 *SCARB1* common HapMap-CEU tagSNPs (**Table A4**); 32 *SCARB1* common HapMap-CEU tagSNPs were selected for genotyping. Enlarged figures: **A**, **B**, and **C** represent bins: 4, 12, and 13, respectively. Detailed RegulomeDB (version 1.0) functions of common HapMap-CEU tagSNPs linked to lipid-associated SNPs (P <0.05) observed in this study are described in **Table A18**. ApoB indicates apolipoprotein B; CEU, Utah residents with northern and western European ancestry; HDL-C, high-density lipoprotein cholesterol; and SNP, single nucleotide polymorphism.

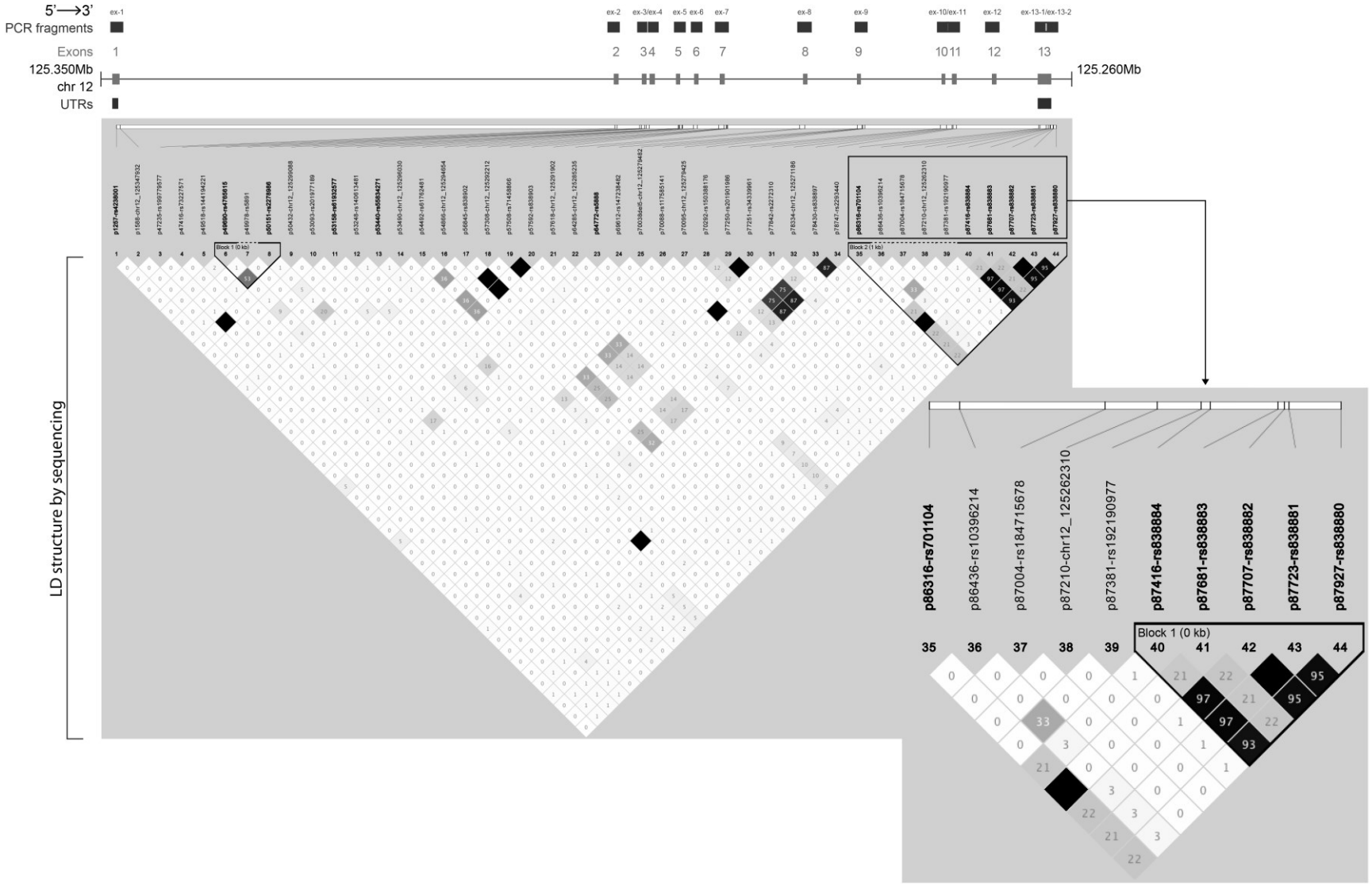


Figure A2. Linkage disequilibrium structure of 44 *SCARB1* sequence variants.

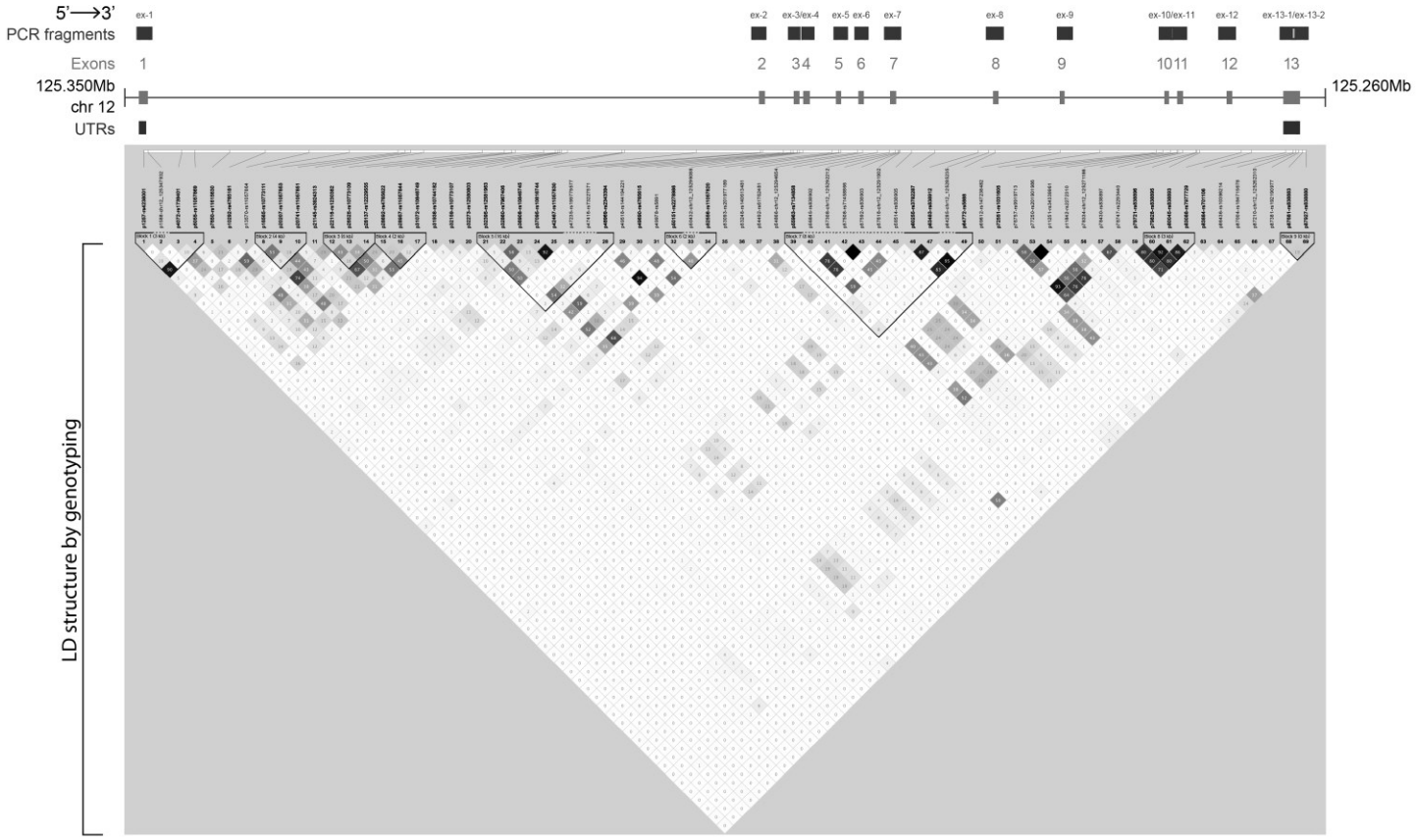


Figure A3. Linkage disequilibrium structure of 69 *SCARB1* genotyped variants.

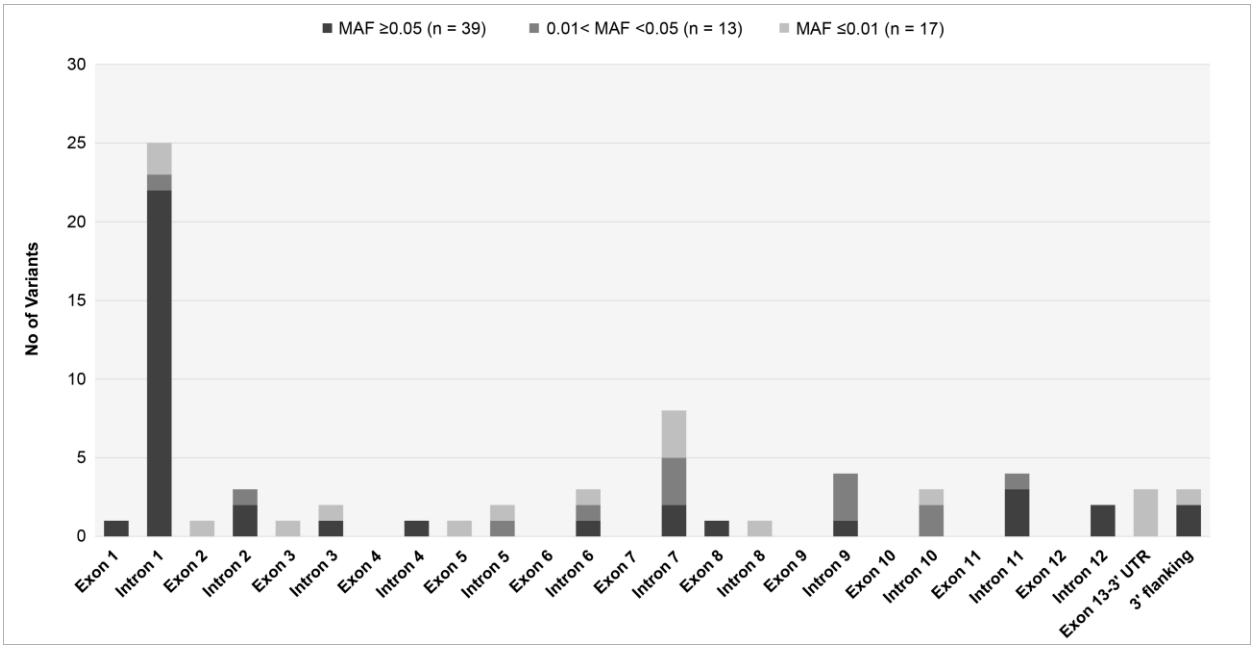
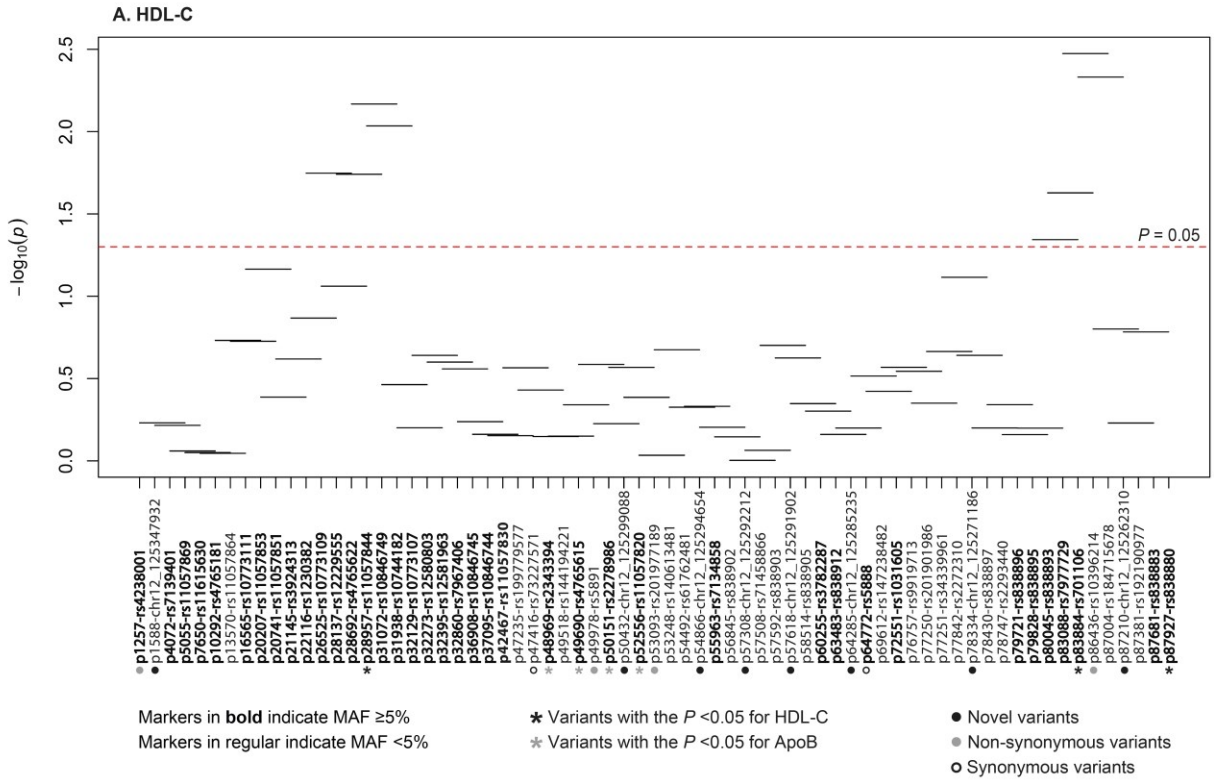


Figure A4. Regional and minor allele frequency (MAF) distributions of 69 *SCARB1* genotyped variants.

Details for each variant are shown in **Table A5**. The *SCARB1* RefSeq (hg19, NM_005505) was obtained from CHIP Bioinformatics. UTR indicates untranslated region.


A. HDL-C
$-\log_{10}(p)$
P = 0.05
Markers in bold indicate MAF ≥5%
Markers in regular indicate MAF <5%
Variants with the P <0.05 for HDL-C
Variants with the P <0.05 for ApoB
Novel variants
Non-synonymous variants
Synonymous variants

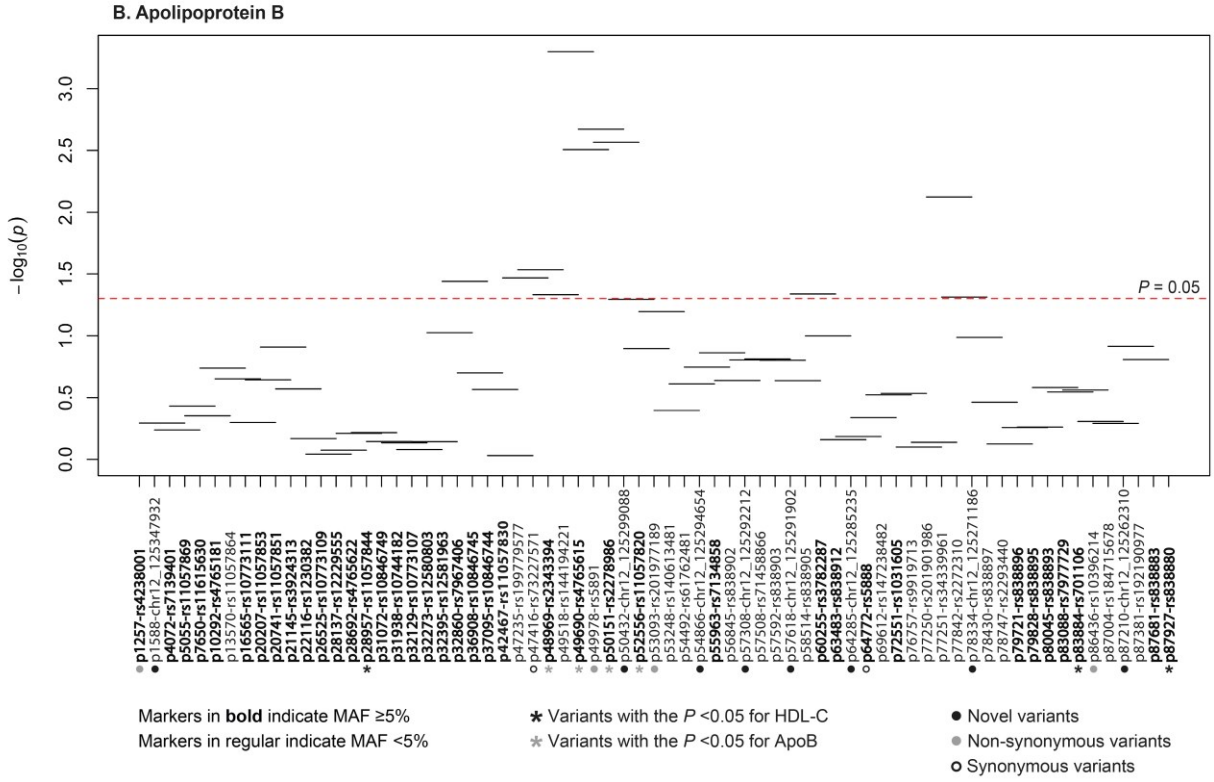
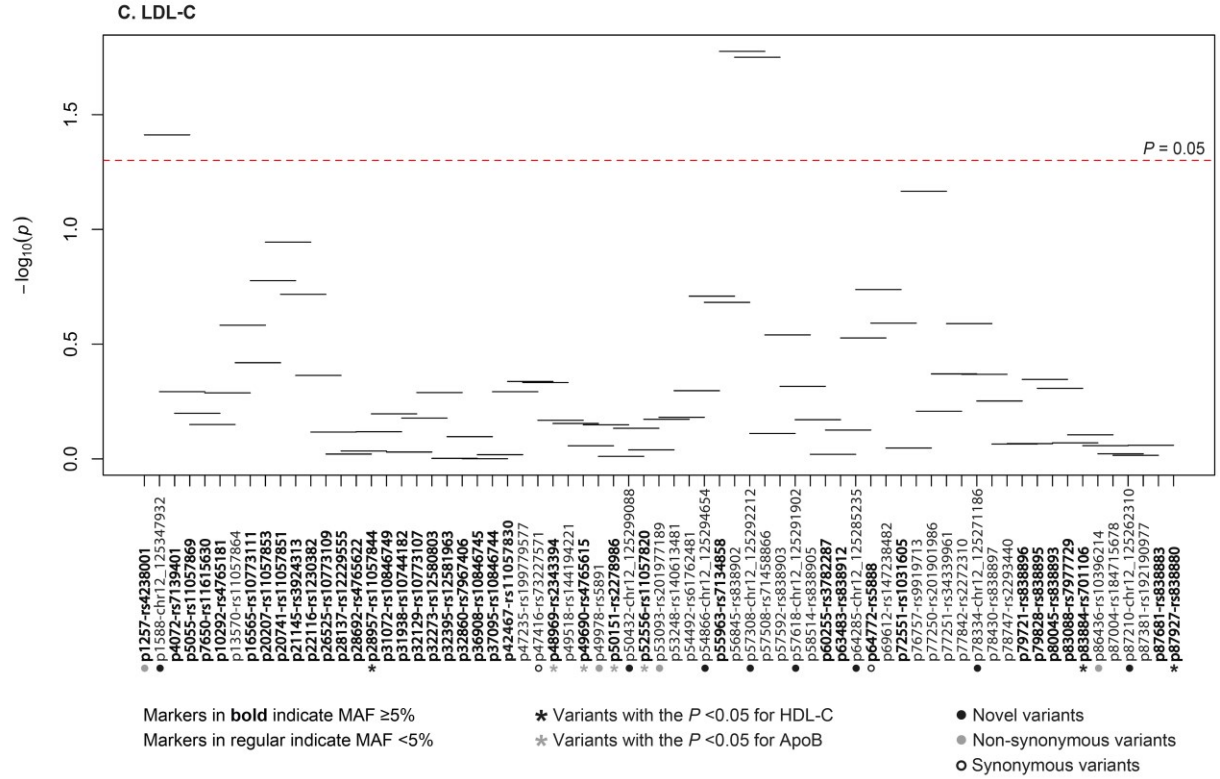

B. Apolipoprotein B
$-\log_{10}(p)$
P = 0.05
Markers in bold indicate MAF ≥5%
Markers in regular indicate MAF <5%
Variants with the P <0.05 for HDL-C
Variants with the P <0.05 for ApoB
Novel variants
Non-synonymous variants
Synonymous variants

Figure A5. (continued)



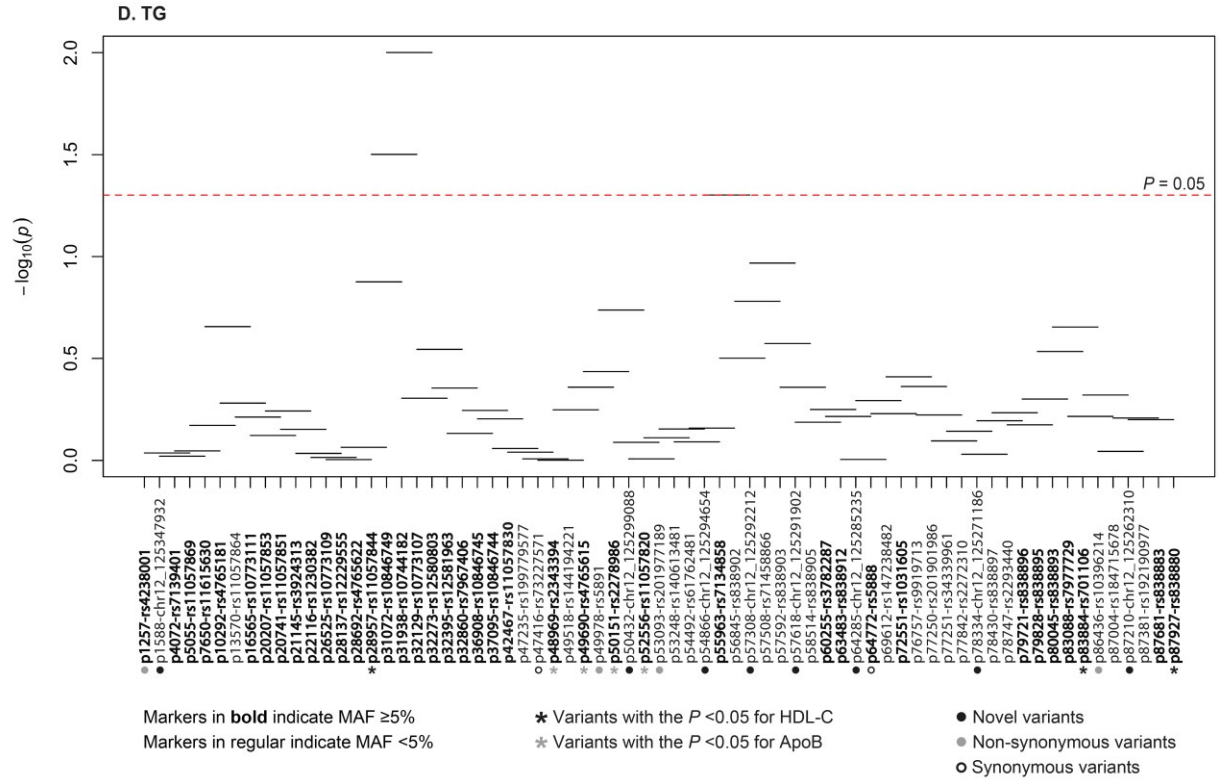


Figure A5. Haplotype association plots of 69 *SCARB1* genotyped variants for HDL-C (A), ApoB (B), LDL-C (C), and TG (D).

The -$\log_{10}$ *P*-values are presented in the Y-axis. A total of 69 genotyped variants are in order of *SCARB1* gene (chromosome 12, 5'→3'; RefSeq: hg19, NM_005505) in the X-axis. Marker names are shown as "SNP name-SNP ID/chromosome 12 position (for novel variants)". SNP IDs are based on dbSNP build 139 (GRCh37.p10); 11 novel variants were submitted to dbSNP database:
http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA). MAF ≥5% are shown in **bold**. Dash line indicates the significance threshold (global *P* = 0.05). ApoB indicates apolipoprotein B; HDL-C, high-density lipoprotein cholesterol; LDL-C, low-density lipoprotein cholesterol; MAF, minor allele frequency; SNP, single nucleotide polymorphism; and TG, triglycerides.

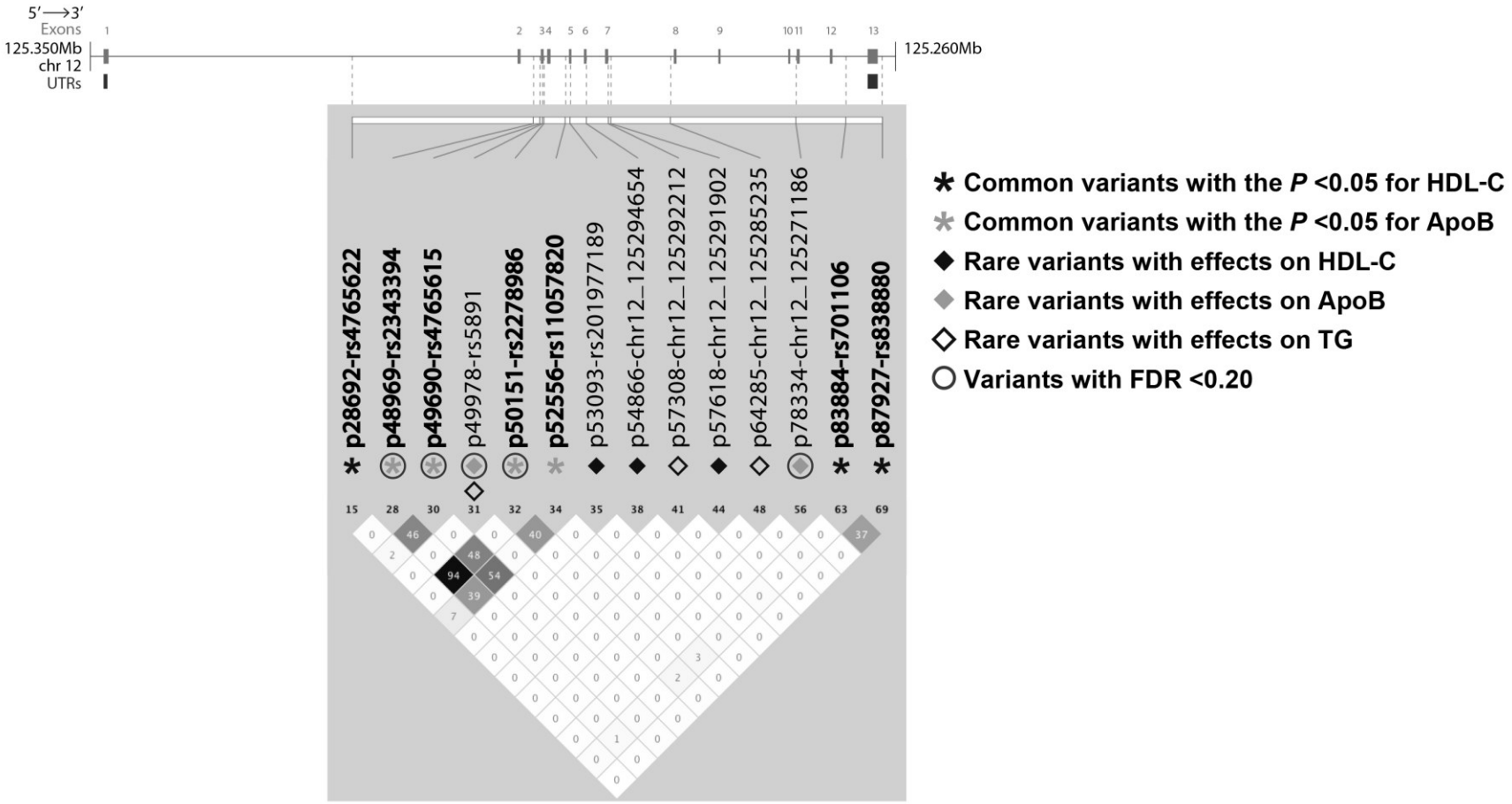


Figure A6. Linkage disequilibrium structure of 14 *SCARB1* variants with single-site *P*-values <0.05.

There are a total of 14 *SCARB1* variants (MAF ≥5%, n = 7; MAF <1%, n = 7) with *P*-values <0.05 observed in single-site analysis (see details in **Tables A8-A11**). Characteristics of 7 common variants (MAF ≥5%) and 7 rare variants (MAF <1%) are summarized in

Table 6.2 and **Table 6.4**, respectively. All 14 variants are in order of *SCARB1* gene (chromosome 12, 5'→3'; RefSeq: hg19, NM_005505) (**top**). Marker names are shown as "SNP name-SNP ID/chromosome 12 position (for novel variants)". SNP IDs are based on dbSNP build 139 (GRCh37.p10); 11 novel variants were submitted to dbSNP database: http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=KAMBOH (batch ID: SCARB1_EA). MAF ≥5% are shown in **bold**. ApoB indicates apolipoprotein B; FDR, false discovery rate; HDL-C, high-density lipoprotein cholesterol; MAF, minor allele frequency; SNP, single nucleotide polymorphism; and TG, triglycerides.