Online Table 1. Primer sequences for *ABCA1* screening.

PCR Fragment	Primer Sequence	PCR Fragment	Primer Sequence	PCR Fragment	Primer Sequence
Core promoter F	5’ caa ctc cct aga tgt gtc gt	Exon 17F	5’ ctc cag gta taa ttt aca cta g	Exon 34F	5’ gaa cct tgt aca cac tcg c
Core promoter R	5’ gct cgc aat tac ggg gtt tt	Exon 17R	5’ tag cac aaa gaa agg aca tca g	Exon 34R	5’ ggt gat ccg ttt aac ctg c
Exon 1F	5’ ggc ttt gac cga tag taa cc	Exon 18F	5’ ggg aaa ccc tca gaa tac tgg g	Exon 35F	5’ tcc tgt tgt cca cag gtt cc
Exon 1R	5’ ttg cgt ctc ttt ctc cta cc	Exon 18R	5’ cac agt ctt tta gaa agg cag gag	Exon 35R	5’ atg ccc ctg cca act tta cc
Exon 2F	5’ agc tga gag ggc tgg att agc	Exon 19F	5’ ctg tgt cct tac act cca ctc c	Exon 36F	5’ aac ggt ctc tgc agc tgt tcc
Exon 2R	5’ ccc tgt gtg aaa acc aac aca gc	Exon 19R	5’ cta act cta ctg cag aac cct cc	Exon 36R	5’ cat ggt tgg tgg gta tgc acc
Exon 3F	5’ att ccc aga tcc cag tgg	Exon 20F	5’ gga gca tct aga gtt aag ggt agg	Exon 37F	5’ gtg tgt acg tgt gtg ttt gtc
Exon 3R	5’ gtg atg ttt ccc aaa gac agc	Exon 20R	5’ taa gtc caa ctc ctc cca tga tgg	Exon 37R	5’ gga aca ttt cct gat gat agc
Exon 4F	5’ gca ttg tat aaa tgg agc c	Exon 21F	5’ ggc cag ggg aca ctg tat tct gg	Exon 38F	5’ ctg ccc ttt ctt gtc tga taa tgg
Exon 4R	5’ cag cca ttc aaa att ctc c	Exon 21R	5’ ttc ctc cgc atg tgt gta gcc gg	Exon 38R	5’ gat ctt gaa cac aaa gga ggc t
Exon 5F	5’ ggc agt cac ttc tgt aaa gca g	Exon 22F	5’ gtc ctc tgc ctt cac tca ctg tct	Exon 39F	5’ gtg gat aag gca ttt ggt gc
Exon 5R	5’ atc taa tgg gaa caa gcc cca g	Exon 22R	5’ ttc tca aaa gcc ccc cgc tct	Exon 39R	5’ taa aca ctg tcc tct ggc tc
Exon 6F	5’ ttc tcc act gtc cca ttg tgc	Exon 23F	5’ gtc cct gtc act gtg aca aat cag	Exon 40F	5’ cca tga atg aca ccc gtt tc
Exon 6R	5’ cac cac cat tac aag gac aag	Exon 23R	5’ tca ttc aca gcc agc aag tcc tg	Exon 40R	5’ gcc atc tcc att aaa gca tcc
Exon 7F	5’ gtt ctc caa aag act tca agg	Exon 24F	5’ cac aac aga gct tct tgg aag cc	Exon 41F	5’ ttg atc cct cca ttc tga c
Exon 7R	5’ caa agt cat gct gtc caa gg	Exon 24R	5’ ttg agt cag cgc cac cag cc	Exon 41R	5’ cat tgg tga gtg ttt ccc tg
Exon 8F	5’ gtt agg agt cgg ttt ctt g	Exon 25F	5’ tcc gaa gcc cct ctg agt tga tt	Exon 42F	5’ cgt tag aga ctg aat ctt tg
Exon 8R	5’ caa ctc aaa agg acc tct tg	Exon 25R	5’ gct act ggt ctg gcc tta gga cat t	Exon 42R	5’ taa gca agt cag caa act gc
Exon 9F	5’ ttc tca tcc cca acc ctt gca c	Exon 26F	5’ ggc agt ggt gac tac tct cgt aa	Exon 43F	5’ ttc cac agt tgc ttg gga ga
Exon 9R	5’ cta cag agg gag gag atg aca c	Exon 26R	5’ gtg cac tga gaa agc cag cag aa	Exon 43R	5’ tgc ttg att ggg tag aga tag
Exon 10F	5’ act ctc tgg ctc caa agc cct tg	Exon 27F	5’ gca gat ggg gaa ggt ttg tt	Exon 44F	5’ gaa ggt ttg agg tag tta cg
Exon 10R	5’ cgc aca cct ctg aag cta cct tg	Exon 27R	5’ aca ggt gag agc atg aga ga	Exon 44R	5’ gaa gag aat gag act gaa agg
Exon 11F	5’ tgg ggt atc ttt tca ggg ag	Exon 28F	5’ gaa gaa gga gca tgg caa tg	Exon 45F	5’ acc aga gta tct cta cct gc
Exon 11R	5’ aga att ctc acc tca ctc aca	Exon 28R	5’ ttc act ggt cac aga gcc tg	Exon 45R	5’ cag cct gaa gtc aat gcg tg
Exon 12F	5’tta cct cag ttt tct gtg agg	Exon 29F	5’ ggc ctt gtt tct aac act tgc	Exon 46F	5’ cat gta tgt gta gga cag ca
Exon 12R	5’ tcc tgt gtt agg ctt gag g	Exon 29R	5’ cct cgt aaa cat ctt tgg tct gc	Exon 46R	5’ gcc ctg gac ata tag gac tt
Exon 13F	5’ atg agc aat cgt gta gtc ag	Exon 30F	5’ gag cct gtc aca gag aaa tg	Exon 47F	5’ gct gtt tca aag atg ctt ctg c
Exon 13R	5’ gta cct ctt gtc cta ata taa tag	Exon 30R	5’ cac aat gtg gca tgc agt tg	Exon 47R	5’ gag cag tat tta tac tgg gct tg
Exon 14F	5’ aag ggt agt agg gtt ttg ggc	Exon 31F	5’ ctc ctg agg caa cag aaa gg	Exon 48F	5’ ggt gat gtt ctc atg gtt ac
Exon 14R	5’ cac aca tgc atg cac atg cac	Exon 31R	5’ tca ttc ctg ctt cca agc gg	Exon 48R	5’ agt tcg gac ttc aaa gcc ct
Exon 15F	5’ atg cag gct cag agg cct tg	Exon 32F	5’ gac ctg tag tca ggt ttc c	Exon 49F	5’ atg gca caa gtt ggg ttc cca
Exon 15R	5’ tcc tcc ctt agc ccg tgt tg	Exon 32R	5’ ggc cat aat ctg gca ttt cc	Exon 49R	5’ gat aca aac tgc tct tgg acc
Exon 16F	5’ tac aag tga gtg ctt ggg at	Exon 33F	5’ cct cag gtg gat att tac tcc	Exon 50F	5’ gta ttc ttc cct cct tca ca
Exon 16R	5’ ctc caa agg aag gtc aaa tg	Exon 33R	5’ tcc aag aca cct aca aca cc	Exon 50R	5’ cac aac act tca cat ggt gca

F=forward, R=reverse

Online Table 2. Genetic variation in introns of *ABCA1* in individuals from the general population with low HDL-C levels.

Gene region	Nucleotide Substitution*	No. of Alleles Low HDL-C (n=190)	References and dbSNP ID numbers
Intron 2	IVS2-50T>C	22	dbSNP 2275545
Intron 3	IVS3+26G>A	2	New
Intron 4	IVS4+64C>T	1	New
Intron 5	IVS5+75T>G	3	New
Intron 8	IVS8-15-(-14)insA	10	dbSNP 3215917
Intron 10	IVS10-62-(-61)insG	37	dbSNP 4149346
Intron 12	IVS12+85-(+86)insT	12	New
Intron 14	IVS14+24T>A	40	dbSNP 4743763
Intron 14	IVS14-59C>T	9	dbSNP 2066717
Intron 23	IVS23-65G>A	3	New
Intron 25	IVS25+23G>A	1	New
Intron 32	IVS32+30T>G	6	dbSNP 2777801
Intron 33	IVS33-26G>C	9	dbSNP 2297404
Intron 36	IVS36-55T>C	1	New
Intron 37	IVS37-47T>C	2	New
Intron 44	IVS44+18T>C	24	dbSNP 2020927
Intron 45	IVS45+68T>C	29	dbSNP 4149336
Intron 46	IVS46-34C>T	4	New
Intron 48	IVS48+13A>G	11	dbSNP 2740485
Intron 48	IVS48+86A>C	20	New
Intron 48	IVS48+117A>G	15	dbSNP 2066882
Intron 49	IVS49+55G>C	29	dbSNP 1331924

*Nucleotide 1 denotes A in the startcodon ATG in exon 2, corresponding to base position 314 in mRNA sequence NM_005502.2. Approximately 100 bp up- and downstream from the splice sites were screened in introns 1-50. HDL-C=high density lipoprotein cholesterol, IVS=intervening sequence.

Online Table 3. Pairwise linkage disequilibrium of *ABCA1* single nucleotide polymorphisms in individuals from the general population with low and high HDL-C levels.

	-413G>C	-328C>T	-279C>G	-75-(-76)insG	-18G>C	L158	R219K	P312	G316	I680	V771M	V825I	I883M	E1172D	T1427	T1512	R1587K
-413G>C		-1.0^{C}	+1.0^{A}	NS	NS	NS	NS	-1.0^{B}	NS	NS	NS	NS	NS	NS	NS	NS	NS
-328C>T	-1.0^{C}		NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS
-279C>G	+1.0^{B}	NS		NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS
-75-(-76)insG	NS	NS	+0.4^{A}		+1.0^{C}	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS
-18G>C	NS	NS	+0.4^{A}	+1.0^{C}		NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS
L158	+0.3^{A}	NS	NS	NS	NS		+0.7^{C}	+0.5^{B}	+0.7^{B}	NS	NS	NS	NS	NS	+0.5^{A}	NS	NS
R219K	+0.3^{A}	NS	NS	NS	NS	+0.7^{C}		+0.8^{C}	+0.8^{B}	NS	NS	NS	+0.4^{A}	NS	+0.6^{A}	NS	NS
P312	NS	NS	+0.4^{A}	NS	NS	+0.5^{B}	+0.8^{C}		NS	NS	NS	NS	NS	+0.5^{A}	NS	NS	NS
G316	NS	NS	NS	NS	NS	+0.6^{C}	+0.9^{C}	NS		NS	+1.0^{A}	NS	NS	NS	NS	NS	NS
I680	NS	NS	+0.4^{A}	NS	NS	NS	NS	NS	-1.0^{A}		NS	+1.0^{C}	+1.0^{C}	NS	+0.7^{C}	NS	NS
V771M	NS	NS	NS	NS	NS	+0.6^{B}	+1.0^{C}	NS	+1.0^{C}	NS		NS	NS	NS	+0.9^{A}	NS	NS
V825I	NS	NS	NS	NS	NS	NS	NS	NS	NS	+1.0^{C}	NS		+1.0^{C}	NS	NS	NS	NS
I883M	NS	NS	+0.4^{A}	NS	NS	NS	NS	NS	-1.0^{A}	+1.0^{C}	NS	+0.9^{C}		NS	+0.7^{C}	NS	NS
E1172D	NS	NS	NS	NS	NS	NS	NS	NS	+0.6^{B}	NS	+0.5^{B}	NS	NS		NS	NS	+1.0^{B}
T1427	NS	-0.8^{A}	NS	NS	NS	+0.3^{A}	+0.4^{A}	NS	NS	+0.5^{C}	NS	+0.3^{A}	+0.5^{C}	NS		NS	-1.0^{A}
T1512	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS	NS		NS
R1587K	NS	NS	+0.6^{A}	NS	NS	NS	+0.5^{B}	+0.9^{C}	NS	NS	NS	NS	NS	+1.0^{C}	-1.0^{A}	NS	

Disequilibrium statistics are reported as D', ranging from –1.0 to +1.0 ($D'=D/D_{max}$ when $D>0$, $D'=D/D_{min}$ when $D<0$). $D=h-pq$, where h is the frequency of the rare estimated haplotype for a pair of sites, p and q are the frequencies, assuming no linkage, of the alleles in that haplotype. D_{max} is $p(1-q)$ and D_{min} is pq, assuming that $p<q<0.5$. + indicates that rare alleles at each locus segregate together. – indicates that the rare allele at one locus segregates with the common allele at the other locus. Estimates in the upper right hand corner are for individuals with low HDL-C and in the lower left hand corner for individuals with high HDL-C levels. For individuals with low HDL-C, the first locus is on the vertical axis and the second locus is on the horizontal axis. For individuals with high HDL-C, the first locus is on the horizontal axis and the second locus is on the vertical axis. $^{A}p<0.05$, $^{B}p<0.01$, $^{C}p<0.001$, NS=not significant.