Locus 208 (rs1801689)

SNiPAcard

Block annotations

Block info

genomic range	chr17:64,195,431-64,305,051
block size	109,621 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 1.057 [-1.088 – 6.439]	gene(s) hit or close-by	APOH, PRKCA, PSMD7P1
phastCons	μ = 0.241 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 1.266 [-2.42 – 5.5]	potentially regulated gene(s)	–
CADD score	μ = 8.965 [0.147 – 26.9]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
LDL cholesterol	<1.00×10-11	GWAS Catalog	24097068	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM970094	1

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
APOH	missense variant	ENST00000205948	NM_000042.2	ENSP00000205948	G/C	Ggc/Tgc	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001537522 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	FOSL2, Jund, PolII, H3K79me2, DNase1, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, H3K27me3, H3K36me3
		lung (IMR90)	H3K27me3
		A549	H3K4me3, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APOH	downstream gene variant, upstream gene variant	20	ENST00000581797	?	ENSP00000463553	3

APOH	upstream gene variant	3460	ENST00000205948	NM_000042.2	ENSP00000205948	1
PRKCA	downstream gene variant	2585	ENST00000583775	?	?	1
PSMD7P1	downstream gene variant	1075	ENST00000579375	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOH	ENST00000205948	NM_000042.2	ENSP00000205948	1
APOH	ENST00000577982	?	ENSP00000464301	2
APOH	ENST00000585162	?	ENSP00000462260	1
PRKCA	ENST00000284384	?	ENSP00000284384	1
PRKCA	ENST00000578063	?	ENSP00000462087	1
PRKCA	ENST00000413366	NM_002737.2	ENSP00000408695	1
PRKCA	ENST00000583361	?	?	1