Locus 212 (rs8178847)

SNiPAcard

Block annotations

Block info

genomic range	chr17:64,190,994-64,235,007
block size	44,014 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.099 [-2.05 – 1.743]	gene(s) hit or close-by	APOH, CEP112, PSMD7P1
phastCons	μ = 0.053 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 0.154 [-2.89 – 3.46]	potentially regulated gene(s)	–
CADD score	μ = 3.663 [0.212 – 9.646]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR033517	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	2	2			2
APOH	missense variant	ENST00000581797	?	ENSP00000463553	2	2			2
APOH	missense variant	ENST00000577982	?	ENSP00000464301	2	2			2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001537522 (promoter)	3	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
ENSR00001537523 (enhancer)	1	HSMMtube	H3K27me3
		liver (HepG2)	H3K27ac, H3K4me2, H3K4me1
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

APOH	downstream gene variant	1109	ENST00000577982	?	ENSP00000464301	5
APOH	downstream gene variant, upstream gene variant	67	ENST00000585162	?	ENSP00000462260	8
APOH	upstream gene variant	197	ENST00000581797	?	ENSP00000463553	4
APOH	downstream gene variant, upstream gene variant	1383	ENST00000205948	NM_000042.2	ENSP00000205948	3
CEP112	upstream gene variant	3007	ENST00000392769	NM_145036.3	ENSP00000376522	2
CEP112	upstream gene variant	3179	ENST00000537949	?	ENSP00000440775	2
CEP112	upstream gene variant	2792	ENST00000580624	?	?	2
CEP112	upstream gene variant	2797	ENST00000535342	NM_001199165.1	ENSP00000442784	2
CEP112	upstream gene variant	3021	ENST00000583358	?	ENSP00000463914	2
PSMD7P1	upstream gene variant, downstream gene variant	1284	ENST00000579375	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOH	ENST00000585162	?	ENSP00000462260	5
APOH	ENST00000581797	?	ENSP00000463553	14
APOH	ENST00000205948	NM_000042.2	ENSP00000205948	15
APOH	ENST00000577982	?	ENSP00000464301	13

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOH	ENST00000205948	NM_000042.2	ENSP00000205948	1
APOH	ENST00000577982	?	ENSP00000464301	1