Locus 53 (rs3138036)

SNiPAcard

Block annotations

Block info

genomic range	chr17:32,647,544-32,660,149
block size	12,606 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.302 [-3.87 – 2.066]	gene(s) hit or close-by	CCL8, RP11-521P1.1
phastCons	μ = 0.079 [0 – 0.941]	eQTL gene(s)	–
GERP++	μ = -0.332 [-5.69 – 3.09]	potentially regulated gene(s)	–
CADD score	μ = 4.854 [0.181 – 22.6]	disease gene(s)	–

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)
CCL8	missense variant	ENST00000394620	NM_005623.2	ENSP00000378118	Q/K	Caa/Aaa	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001638184 (CTCF binding site)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		A549	H3K27me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL8	downstream gene variant	1316	ENST00000394620	NM_005623.2	ENSP00000378118	9
RP11-521P1.1	downstream gene variant, upstream gene variant	902	ENST00000582638	?	?	8

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL8	ENST00000394620	NM_005623.2	ENSP00000378118	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-521P1.1	ENST00000582638	?	1