Locus 428 (rs35931837)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,033,158-52,033,158
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.302	gene(s) hit or close-by	SIGLEC6
phastCons	0.083	eQTL gene(s)	SIGLEC12
GERP++	1.55	potentially regulated gene(s)	–
CADD score	8.342	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)
SIGLEC6	missense variant	ENST00000425629	NM_001245.5	ENSP00000401502	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000359982	NM_001177548.1	ENSP00000353071	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000436458	NM_001177547.1	ENSP00000410679	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000343300	NM_198846.4	ENSP00000345907	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000391797	NM_001177549.1	ENSP00000375674	S/G	Agc/Ggc	?	?	1
SIGLEC6	missense variant	ENST00000346477	NM_198845.4	ENSP00000344064	S/G	Agc/Ggc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC12	?	ENSG00000254521	blood	5.91×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SIGLEC6	upstream gene variant	1457	ENST00000474054	?	?	1

Putative effect on transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SIGLEC6	ENST00000489837	?	1
SIGLEC6	ENST00000496422	?	1

Locus 428 (sentinel SNP rs35931837 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.