Locus 223 (rs10421385)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,677,669-51,704,168
block size	26,500 bp
variant count	28 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.878 [-4.83 – 0.386]	gene(s) hit or close-by	CTD-3187F8.14, CTD-3187F8.7, SIGLEC17P, SIGLEC20P, SIGLEC21P
phastCons	μ = 0.074 [0 – 0.973]	eQTL gene(s)	SIGLEC20P
GERP++	μ = -0.931 [-5.39 – 1.22]	potentially regulated gene(s)	–
CADD score	μ = 2.421 [0.144 – 14.52]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC20P	?	ENSG00000268336	testis	7.34×10^{-6} (p-value)	GTEx Portal V6	6

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001649028 (open chromatin region)	3	embryonic stem cell (H1ESC)	DNase1
		A549	H3K27me3
		blood (K562)	H3K27me3
ENSR00000217706 (open chromatin region)	1	A549	H3K27me3
		blood (K562)	DNase1, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-3187F8.14	upstream gene variant	1388	ENST00000600074	?	?	9
CTD-3187F8.7	downstream gene variant, upstream gene variant	645	ENST00000595913	?	?	12
SIGLEC17P	downstream gene variant	984	ENST00000614626	?	?	2
SIGLEC17P	downstream gene variant	889	ENST00000618545	?	?	2
SIGLEC17P	downstream gene variant	891	ENST00000619985	?	?	2
SIGLEC17P	downstream gene variant	913	ENST00000611992	?	?	2
SIGLEC20P	upstream gene variant, downstream gene variant	35	ENST00000595243	?	?	11
SIGLEC21P	upstream gene variant	220	ENST00000598990	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-3187F8.14	ENST00000600074	?	?	2
CTD-3187F8.7	ENST00000595913	?	?	1

SIGLEC20P	ENST00000595243	?	?	10
SIGLEC21P	ENST00000598990	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SIGLEC20P	ENST00000595243	?	2