Locus 136 (rs2005199)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,016,530-52,033,572
block size	17,043 bp
variant count	28 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.517 [-3.498 – 0.854]	gene(s) hit or close-by	CTD-3073N11.9, SIGLEC27P, SIGLEC6
phastCons	μ = 0.077 [0 – 0.626]	eQTL gene(s)	LRRC4B, ZNF175
GERP++	μ = -0.741 [-3.21 – 1.89]	potentially regulated gene(s)	SIGLEC10, SIGLEC12, SIGLEC14, SIGLEC6
CADD score	μ = 3.671 [0.042 – 16.32]	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	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000346477	NM_198845.4	ENSP00000344064	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000436458	NM_001177547.1	ENSP00000410679	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000343300	NM_198846.4	ENSP00000345907	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000391797	NM_001177549.1	ENSP00000375674	F/L	Ttc/Ctc	?	?	1
SIGLEC6	missense variant	ENST00000359982	NM_001177548.1	ENSP00000353071	F/L	Ttc/Ctc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ZNF175	ENST00000436511	ILMN_1675788	blood	4.95×10-7 (p-value)	MuTHER consortium	12
ZNF175	ENST00000262259					
ZNF175	?	ENSG00000105497	esophagus mucosa	2.20×10-6 (p-value)	GTEx Portal V6	4
LRRC4B	?	ENSG00000131409	lung	1.70×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000342383	1	ENST00000425629, ENST00000436458, ENST00000391797, ENST00000346477, ENST00000359982	SIGLEC6

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000232795	1	ENCP00000024652	SIGLEC14

	ENCP00000024646	SIGLEC6
	ENCP00000024642	SIGLEC10
	ENCP00000024643	SIGLEC10
	ENCP00000024645	SIGLEC12

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000217752 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	CTCF, H3K27me3
		blood (K562)	CTCF, Rad21, DNase1
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-3073N11.9	upstream gene variant, downstream gene variant	220	ENST00000598220	?	?	16
SIGLEC27P	downstream gene variant	3965	ENST00000594477	?	?	1
SIGLEC6	downstream gene variant	14	ENST00000346477	NM_198845.4	ENSP00000344064	8
SIGLEC6	downstream gene variant, upstream gene variant	225	ENST00000474054	?	?	11
SIGLEC6	downstream gene variant	406	ENST00000489837	?	?	7
SIGLEC6	downstream gene variant	219	ENST00000436458	NM_001177547.1	ENSP00000410679	6
SIGLEC6	downstream gene variant	488	ENST00000391797	NM_001177549.1	ENSP00000375674	6
SIGLEC6	downstream gene variant	219	ENST00000343300	NM_198846.4	ENSP00000345907	6
SIGLEC6	downstream gene variant	219	ENST00000359982	NM_001177548.1	ENSP00000353071	6
SIGLEC6	downstream gene variant	236	ENST00000425629	NM_001245.5	ENSP00000401502	6
SIGLEC6	downstream gene variant	277	ENST00000496422	?	?	7

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC6	ENST00000489837	?	?	2
SIGLEC6	ENST00000436458	NM_001177547.1	ENSP00000410679	18
SIGLEC6	ENST00000346477	NM_198845.4	ENSP00000344064	18
SIGLEC6	ENST00000391797	NM_001177549.1	ENSP00000375674	18
SIGLEC6	ENST00000425629	NM_001245.5	ENSP00000401502	18
SIGLEC6	ENST00000474054	?	?	14
SIGLEC6	ENST00000359982	NM_001177548.1	ENSP00000353071	18

SIGLEC6	ENST00000343300	NM_198846.4	ENSP00000345907	18
SIGLEC6	ENST00000496422	?	?	2

Non-coding exon variant

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