SNiPAcard

Block annotations

Block info

genomic range	chr19:51,632,705-51,636,001
block size	3,297 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.624 [-2.525 – 0.424]	**gene(s) hit or close-by**	SIGLEC9
phastCons	μ = 0.035 [0.001 – 0.103]	**eQTL gene(s)**	–
GERP++	μ = -0.323 [-0.764 – 0.113]	**potentially regulated gene(s)**	–
CADD score	μ = 3.481 [0.49 – 7.265]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001649020 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K4me3
		embryonic stem cell (H1ESC)	CTCF
		endothelium (HUVEC)	H3K36me3, DNase1
		blood (K562)	CTCF, DNase1
		A549	H3K27me3
ENSR00000217699 (promoter flanking region)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1
		embryonic stem cell (H1ESC)	CTCF
		A549	H3K27me3
		blood (K562)	CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SIGLEC9	downstream gene variant	490	ENST00000599948	?	ENSP00000472483	3
SIGLEC9	downstream gene variant	93	ENST00000250360	NM_014441.2	ENSP00000250360	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC9	ENST00000440804	NM_001198558.1	ENSP00000413861	3
SIGLEC9	ENST00000250360	NM_014441.2	ENSP00000250360	1