Locus 39 (rs7250849)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,151,087-52,160,765
block size	9,679 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.381 [-1.801 – -0.878]	**gene(s) hit or close-by**	RPL7P51, SIGLEC14, SIGLEC5, SIGLEC5
phastCons	μ = 0.002 [0 – 0.006]	**eQTL gene(s)**	RPL7P51, SIGLEC14, SIGLEC5
GERP++	μ = -1.326 [-2 – -0.513]	**potentially regulated gene(s)**	–
CADD score	μ = 1.757 [0.015 – 5.867]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC14	?	ENSG00000254415	visceral adipocytes	7.21×10^{-9} (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	muscularis mucosae	1.95×10^{-13} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	lung	1.95×10^{-13} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	tibial nerve	2.48×10^{-16} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	sigmoid colon	1.04×10^{-8} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	atrial appendage	9.58×10^{-10} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	tibial artery	1.27×10^{-13} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	blood	7.77×10^{-16} (p-value)	GTEx Portal V6	4
SIGLEC5	?	ENSG00000105501	blood	8.62×10^{-11} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	thyroid	1.55×10^{-14} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	gastroesophageal junction	1.81×10^{-10} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	testis	1.18×10^{-8} (p-value)	GTEx Portal V6	4
SIGLEC5	?	ENSG00000105501	testis	6.00×10^{-6} (p-value)	GTEx Portal V6	1
SIGLEC14	?	ENSG00000254415	sun exposed skin	3.17×10^{-10} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	aorta	6.40×10^{-10} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	left ventricle	5.61×10^{-9} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	subcutaneous adipocytes	2.87×10^{-15} (p-value)	GTEx Portal V6	4
SIGLEC14	?	ENSG00000254415	breast	3.54×10^{-9} (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	skeletal muscle	3.82×10^{-8} (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	EBV lymphocytes	2.75×10^{-8} (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	transverse colon	1.84×10^{-8} (p-value)	GTEx Portal V6	3

SIGLEC5	?	ENSG00000105501	blood	4.75×10-2 (q-value)	SeeQTL DB (HapMap)	1
SIGLEC14	?	ENSG00000254415	caudate basal ganglia	6.53×10-8 (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	spleen	4.15×10-9 (p-value)	GTEx Portal V6	3
RPL7P51	?	ENSG00000243469	spleen	3.14×10-7 (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	cortex	6.04×10-7 (p-value)	GTEx Portal V6	1
SIGLEC14	?	ENSG00000254415	frontal cortex	4.23×10-9 (p-value)	GTEx Portal V6	3
SIGLEC14	?	ENSG00000254415	pancreas	5.09×10-7 (p-value)	GTEx Portal V6	2
SIGLEC14	?	ENSG00000254415	esophagus mucosa	4.64×10-8 (p-value)	GTEx Portal V6	1
RPL7P51	?	ENSG00000243469	nucleus accumbens	1.57×10-7 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000641970 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	CTCF, Rad21
ENSR00000199573 (CTCF binding site)	2	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	CTCF, DNase1
		blood (K562)	Max, DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		blood (GM12878)	DNase1, Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	DNase1, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPL7P51	upstream gene variant	1028	ENST00000490532	?	?	1
SIGLEC14	upstream gene variant	1033	ENST00000360844	NM_001098612.1	ENSP00000354090	1
SIGLEC14	upstream gene variant	2257	ENST00000533866	?	?	1
SIGLEC5	upstream gene variant	2289	ENST00000534261	?	ENSP00000473238	1
SIGLEC5	upstream gene variant	1034	ENST00000599649	?	ENSP00000470259	1
SIGLEC5	upstream gene variant	945	ENST00000429354	?	ENSP00000415200	1