Locus 84 (rs2124910)

SNiPAcard

Block annotations

Block info

genomic range	chr19:52,021,247-52,039,859
block size	18,613 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.322 [-4.185 – 0.489]	gene(s) hit or close-by	CTD-3073N11.9, SIGLEC6
phastCons	μ = 0.097 [0 – 0.973]	eQTL gene(s)	SIGLEC12, SIGLEC5, SIGLEC5, SIGLEC6, ZNF175
GERP++	μ = -1.080 [-5.25 – 1.25]	potentially regulated gene(s)	SIGLEC6
CADD score	μ = 4.196 [0.562 – 11.6]	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	ENST00000359982	NM_001177548.1	ENSP00000353071	P/S	Cca/Tca	?	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SIGLEC5	ENST00000534261	ILMN_1740298	blood	2.78×10-8 (p-value)	MuTHER consortium	13
SIGLEC5	ENST00000599649					
SIGLEC5	ENST00000570106					
SIGLEC5	ENST00000429354					
ZNF175	ENST00000436511	ILMN_1675788	blood	1.07×10-5 (p-value)	MuTHER consortium	12
ZNF175	ENST00000262259					
SIGLEC12	?	ENSG00000254521	pancreas	1.93×10-9 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	muscularis mucosae	5.37×10-19 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	lung	3.79×10-31 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	tibial artery	2.23×10-22 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	breast	9.34×10-13 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	blood	3.73×10-35 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	thyroid	1.67×10-10 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	transverse colon	1.09×10-12 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	sun exposed skin	2.23×10-18 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	aorta	3.18×10-17 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	left ventricle	2.15×10-8 (p-value)	GTEx Portal V6	15
SIGLEC12	?	ENSG00000254521	terminal ileum	3.61×10-9 (p-value)	GTEx Portal V6	15

SIGLEC12	?	ENSG00000254521	subcutaneous adipocytes	3.94×10-24 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	visceral adipocytes	8.50×10-8 (p-value)	GTEx Portal V6	15
SIGLEC12	?	ENSG00000254521	stomach	4.48×10-14 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	spleen	5.33×10-16 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	tibial nerve	3.18×10-16 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	coronary artery	4.81×10-7 (p-value)	GTEx Portal V6	9
SIGLEC12	?	ENSG00000254521	vagina	4.25×10-8 (p-value)	GTEx Portal V6	14
SIGLEC12	?	ENSG00000254521	esophagus mucosa	7.00×10-10 (p-value)	GTEx Portal V6	21
SIGLEC5	ENST00000534261	ILMN_1740298	blood	1.47×10-5 (p-value)	Westra et al.	9
SIGLEC5	ENST00000599649					
SIGLEC5	ENST00000570106					
SIGLEC5	ENST00000429354					
SIGLEC6	ENST00000425629	ILMN_2299095	blood	8.57×10-5 (p-value)	Westra et al.	6
SIGLEC6	ENST00000436458					
SIGLEC6	ENST00000391797					
SIGLEC6	ENST00000343300					
SIGLEC6	ENST00000474054					
SIGLEC6	ENST00000346477					
SIGLEC6	ENST00000359982					
SIGLEC12	?	ENSG00000254521	gastroesophageal junction	2.43×10-10 (p-value)	GTEx Portal V6	21
SIGLEC12	?	ENSG00000254521	adrenal gland	4.48×10-13 (p-value)	GTEx Portal V6	21
SIGLEC5	?	ENSG00000105501	blood	4.65×10-2 (q-value)	SeeQTL DB (HapMap)	1
SIGLEC12	ENST00000291707	ILMN_1676363	monocyte	1.17×10-115 (p-value)	Zeller et al.	3
SIGLEC12	ENST00000596742					
SIGLEC12	ENST00000598614					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024646		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001419955 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	Max, PU1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		endothelium (HUVEC)	H3K27me3
		blood (GM12878)	PU1, H3K4me2
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-3073N11.9	upstream gene variant, downstream gene variant	1448	ENST00000598220	?	?	7
SIGLEC6	downstream gene variant, upstream gene variant	646	ENST00000436458	NM_001177547.1	ENSP00000410679	9
SIGLEC6	downstream gene variant, upstream gene variant	646	ENST00000391797	NM_001177549.1	ENSP00000375674	9
SIGLEC6	downstream gene variant, upstream gene variant	105	ENST00000489837	?	?	12
SIGLEC6	downstream gene variant, upstream gene variant	655	ENST00000359982	NM_001177548.1	ENSP00000353071	9
SIGLEC6	downstream gene variant, upstream gene variant	967	ENST00000474054	?	?	7
SIGLEC6	downstream gene variant, upstream gene variant	646	ENST00000425629	NM_001245.5	ENSP00000401502	9
SIGLEC6	downstream gene variant, upstream gene variant	732	ENST00000346477	NM_198845.4	ENSP00000344064	9
SIGLEC6	downstream gene variant, upstream gene variant	593	ENST00000343300	NM_198846.4	ENSP00000345907	9
SIGLEC6	downstream gene variant, upstream gene variant	646	ENST00000496422	?	?	11

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC6	ENST00000496422	?	?	1
SIGLEC6	ENST00000391797	NM_001177549.1	ENSP00000375674	1
SIGLEC6	ENST00000425629	NM_001245.5	ENSP00000401502	1
SIGLEC6	ENST00000343300	NM_198846.4	ENSP00000345907	1
SIGLEC6	ENST00000489837	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC6	ENST00000436458	NM_001177547.1	ENSP00000410679	11
SIGLEC6	ENST00000346477	NM_198845.4	ENSP00000344064	11
SIGLEC6	ENST00000391797	NM_001177549.1	ENSP00000375674	11
SIGLEC6	ENST00000425629	NM_001245.5	ENSP00000401502	11
SIGLEC6	ENST00000474054	?	?	9
SIGLEC6	ENST00000359982	NM_001177548.1	ENSP00000353071	10
SIGLEC6	ENST00000343300	NM_198846.4	ENSP00000345907	11

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIGLEC6	ENST00000436458	NM_001177547.1	ENSP00000410679	1
SIGLEC6	ENST00000346477	NM_198845.4	ENSP00000344064	1
SIGLEC6	ENST00000391797	NM_001177549.1	ENSP00000375674	1
SIGLEC6	ENST00000425629	NM_001245.5	ENSP00000401502	1
SIGLEC6	ENST00000359982	NM_001177548.1	ENSP00000353071	1
SIGLEC6	ENST00000343300	NM_198846.4	ENSP00000345907	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SIGLEC6	ENST00000496422	?	2

SIGLEC6	ENST00000474054	?	1
SIGLEC6	ENST00000489837	?	1

Locus 84 (sentinel SNP rs2124910 + 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.