SNiPAcard

Block annotations

Block info

genomic range	chr1:160,759,519-160,767,737
block size	8,219 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.349 [-1.062 – 0.48]	**gene(s) hit or close-by**	LY9
phastCons	μ = 0.006 [0 – 0.034]	**eQTL gene(s)**	LY9, SLAMF7
GERP++	μ = -0.668 [-2.19 – 0.577]	**potentially regulated gene(s)**	–
CADD score	μ = 4.528 [0.201 – 10.73]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SLAMF7	ENST00000368043	ILMN_1710923	blood	1.14×10^{-7} (p-value)	MuTHER consortium	2
SLAMF7	ENST00000368042					
SLAMF7	ENST00000458602					
SLAMF7	ENST00000444090					
SLAMF7	ENST00000441662					
SLAMF7	ENST00000621377					
SLAMF7	ENST00000359331					
SLAMF7	ENST00000484221					
SLAMF7	ENST00000458104					
LY9	ENST00000392203	ILMN_1731928	b-cell	1.91×10^{-5} (p-value)	Fairfax et al.	1
LY9	ENST00000263285					
LY9	ENST00000368037					
LY9	ENST00000368035					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000167128 (open chromatin region)	1	blood (GM12878)	H3K4me1, DNase1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		blood (K562)	H3K27me3
		breast (HMEC)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LY9	upstream gene variant	1165	ENST00000368037	NM_001261456.1	ENSP00000357016	4
LY9	upstream gene variant	1302	ENST00000485624	?	?	4
LY9	upstream gene variant	1203	ENST00000392203	NM_001261457.1	ENSP00000376039	4
LY9	upstream gene variant	1197	ENST00000474998	?	ENSP00000476321	4
LY9	upstream gene variant	1299	ENST00000471816	?	?	4
LY9	upstream gene variant	1290	ENST00000480837	?	?	4
LY9	upstream gene variant	1278	ENST00000368039	NM_001033667.2	ENSP00000357018	4

LY9	upstream gene variant	1291	ENST00000490902	?	ENSP00000473643	4
LY9	upstream gene variant	1249	ENST00000263285	NM_002348.3	ENSP00000263285	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
LY9	ENST00000474998	?	ENSP00000476321	S	tcT/tcC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LY9	ENST00000368037	NM_001261456.1	ENSP00000357016	2
LY9	ENST00000485624	?	?	2
LY9	ENST00000392203	NM_001261457.1	ENSP00000376039	2
LY9	ENST00000474998	?	ENSP00000476321	2
LY9	ENST00000471816	?	?	2
LY9	ENST00000480837	?	?	2
LY9	ENST00000368039	NM_001033667.2	ENSP00000357018	2
LY9	ENST00000490902	?	ENSP00000473643	2
LY9	ENST00000263285	NM_002348.3	ENSP00000263285	2