Locus 63 (rs489286)

SNiPAcard

Block annotations

Block info

genomic range	chr1:160,710,157-160,725,854
block size	15,698 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.419 [-0.939 – 0.166]	gene(s) hit or close-by	SLAMF7
phastCons	μ = 0.002 [0 – 0.018]	eQTL gene(s)	LY9, SLAMF7
GERP++	μ = -1.916 [-4.78 – 0.85]	potentially regulated gene(s)	B4GALT3, LY9, NCSTN, PVRL4, RP11-544M22.3, SLAMF1
CADD score	μ = 4.046 [0.885 – 11.26]	disease gene(s)	NCSTN, PVRL4

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
NCSTN	ACNE INVERSA, FAMILIAL, 1	OMIM	MIM:142690
PVRL4	ECTODERMAL DYSPLASIA-SYNDACTYLY SYNDROME 1	OMIM	MIM:613573
PVRL4	ECTODERMAL DYSPLASIA-SYNDACTYLY SYNDROME 1	DECIPHER	MIM:613573
PVRL4	Ectodermal dysplasia - syndactyly syndrome	OrphaNet	OrphaNet:247820

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SLAMF7	ENST00000368043	ILMN_1710923	blood	1.56×10^{-15} (p-value)	MuTHER consortium	9
SLAMF7	ENST00000368042					
SLAMF7	ENST00000458602					
SLAMF7	ENST00000444090					
SLAMF7	ENST00000441662					
SLAMF7	ENST00000621377					
SLAMF7	ENST00000359331					
SLAMF7	ENST00000484221					
SLAMF7	ENST00000458104					
SLAMF7	?	ENSG00000026751	blood	5.58×10^{-5} (q-value)	SeeQTL DB (HapMap)	6
LY9	ENST00000392203	ILMN_1731928	b-cell	1.04×10^{-4} (p-value)	Fairfax et al.	1
LY9	ENST00000263285					
LY9	ENST00000368037					
LY9	ENST00000368035					
LY9	ENST00000490902	ILMN_1812278	blood	3.44×10^{-6} (p-value)	Westra et al.	1
LY9	ENST00000368039					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
SLAMF7	ENST00000368043	222838_at	chr1	blood	2.70×10^{-11} (p-value)	Dixon et al.	3
SLAMF7	ENST00000368042						
SLAMF7	ENST00000458602						

SLAMF7	ENST00000444090						
SLAMF7	ENST00000621377						
SLAMF7	ENST00000441662						
SLAMF7	ENST00000359331						
SLAMF7	ENST00000484221						
SLAMF7	ENST00000458104						
LY9	ENST00000368039	231124_x_at	chr1	blood	9.00×10^{-8} (p-value)	Dixon et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000040800	1	ENCP00000005127	NCSTN
		ENCP00000005137	SLAMF1
		ENCP00000005188	B4GALT3
ENCE00000041028	1	ENCP00000005160	RP11-544M22.3
		ENCP00000005170	PVRL4
		ENCP00000005147	LY9

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000671302 (promoter)	3	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	H3K27me3
		blood (GM12878)	TAF1, PU1, PolII, DNase1, Yy1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		blood (K562)	H3K27me3
		skin (NHEK)	DNase1, H3K4me3
ENSR00001587157 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K4me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	H3K27me3
		blood (GM12878)	PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		blood (K562)	H3K27me3
ENSR00001587158 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (GM12878)	PolII, H3K79me2, H3K36me3
		A549	CTCF
		blood (K562)	H3K27me3, CTCF
		skin (NHEK)	DNase1, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SLAMF7	downstream gene variant	1250	ENST00000441662	NM_001282589.1	ENSP00000405605	1
SLAMF7	downstream gene variant	2396	ENST00000488819	?	?	2
SLAMF7	upstream gene variant, downstream gene variant	1243	ENST00000484221	?	?	3
SLAMF7	downstream gene variant	514	ENST00000495334	?	ENSP00000473590	5
SLAMF7	downstream gene variant	1243	ENST00000359331	NM_001282596.1, NM_001282592.1	ENSP00000352281	1
SLAMF7	downstream gene variant	1243	ENST00000368043	NM_021181.4	ENSP00000357022	1
SLAMF7	downstream gene variant	1250	ENST00000444090	NM_001282588.1	ENSP00000416592	1
SLAMF7	downstream gene variant	1250	ENST00000458602	NM_001282591.1	ENSP00000409965	1

SLAMF7	downstream gene variant	1250	ENST00000621377	NM_001282594.1	ENSP00000483774	1
SLAMF7	downstream gene variant	1243	ENST00000368042	NM_001282590.1, NM_001282595.1	ENSP00000357021	1
SLAMF7	downstream gene variant	1250	ENST00000458104	NM_001282593.1	ENSP00000403294	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLAMF7	ENST00000359331	NM_001282596.1, NM_001282592.1	ENSP00000352281	9
SLAMF7	ENST00000368043	NM_021181.4	ENSP00000357022	9
SLAMF7	ENST00000488819	?	?	7
SLAMF7	ENST00000458602	NM_001282591.1	ENSP00000409965	9
SLAMF7	ENST00000458104	NM_001282593.1	ENSP00000403294	9
SLAMF7	ENST00000368042	NM_001282590.1, NM_001282595.1	ENSP00000357021	9
SLAMF7	ENST00000441662	NM_001282589.1	ENSP00000405605	9
SLAMF7	ENST00000444090	NM_001282588.1	ENSP00000416592	9
SLAMF7	ENST00000495334	?	ENSP00000473590	8
SLAMF7	ENST00000621377	NM_001282594.1	ENSP00000483774	9
SLAMF7	ENST00000484221	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLAMF7	ENST00000359331	NM_001282596.1, NM_001282592.1	ENSP00000352281	3
SLAMF7	ENST00000368043	NM_021181.4	ENSP00000357022	3
SLAMF7	ENST00000458602	NM_001282591.1	ENSP00000409965	3
SLAMF7	ENST00000458104	NM_001282593.1	ENSP00000403294	3
SLAMF7	ENST00000368042	NM_001282590.1, NM_001282595.1	ENSP00000357021	3
SLAMF7	ENST00000441662	NM_001282589.1	ENSP00000405605	3
SLAMF7	ENST00000444090	NM_001282588.1	ENSP00000416592	3
SLAMF7	ENST00000621377	NM_001282594.1	ENSP00000483774	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SLAMF7	ENST00000484221	?	4