SNiPAcard

Block annotations

Block info

genomic range	chr11:124,619,407-124,620,147
block size	741 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.805 [-1.378 – -0.232]	**gene(s) hit or close-by**	ESAM, NRGN, RP11-677M14.2, VSIG2
phastCons	μ = 0.000 [0 – 0]	**eQTL gene(s)**	MSANTD2, VSIG2
GERP++	μ = 0.020 [-2.81 – 2.85]	**potentially regulated gene(s)**	–
CADD score	μ = 6.803 [3.496 – 10.11]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
VSIG2	?	ENSG00000019102	transformed fibroblasts	9.20×10-6 (p-value)	GTEx Portal V6	2
MSANTD2	ENST00000526629	ILMN_1773148	monocyte	2.65×10-4 (p-value)	Fairfax et al.	1
MSANTD2	ENST00000533514					
MSANTD2	ENST00000374979					
MSANTD2	ENST00000239614					
MSANTD2	ENST00000524950					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000570678 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me1
		endothelium (HUVEC)	Cjun, H3K4me1, PolII, DNase1, Cmyc
		liver (HepG2)	H3K27me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ESAM	downstream gene variant	2948	ENST00000464067	?	?	2
ESAM	downstream gene variant	3994	ENST00000435477	?	ENSP00000415893	2
ESAM	downstream gene variant	2332	ENST00000444566	?	ENSP00000406689	2
ESAM	downstream gene variant	2872	ENST00000485116	?	?	2
ESAM	downstream gene variant	2872	ENST00000417453	?	ENSP00000389235	2
ESAM	downstream gene variant	2872	ENST00000278927	NM_138961.2	ENSP00000278927	2
NRGN	downstream gene variant	2301	ENST00000284292	NM_001126181.1, NM_006176.2	ENSP00000284292	2
NRGN	downstream gene variant	2305	ENST00000412681	?	ENSP00000399591	2

RP11-677M14.2	upstream gene variant	3174	ENST00000531241	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
VSIG2	ENST00000403470	?	ENSP00000385013	2
VSIG2	ENST00000326621	NM_014312.3	ENSP00000318684	2