Locus 49 (rs2691273)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,596,136-51,600,921
block size	4,786 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.365 [-0.94 – 0.209]	gene(s) hit or close-by	CTU1
phastCons	μ = 0.000 [0 – 0]	eQTL gene(s)	CTU1
GERP++	μ = -4.200 [-4.53 – -3.87]	potentially regulated gene(s)	–
CADD score	μ = 1.494 [1.32 – 1.667]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CTU1	?	ENSG00000142544	thyroid	1.75×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000143070 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K36me3
		blood (K562)	CTCFL, Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K36me3, CTCF
		lung (IMR90)	H3K36me3, CTCF
		blood (GM12878)	DNase1, Yy1, CTCF, H3K36me3
		nervous (NH-A)	CTCF
		skin (NHEK)	H3K4me1, CTCF, H3K36me3, DNase1
		NHLF	CTCF
		Osteobl	CTCF, H3K36me3
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	DNase1, CTCF
		cervix (HeLa-S3)	H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, PolII
		A549	H3K36me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTU1	downstream gene variant	4727	ENST00000421832	NM_145232.3	ENSP00000390011	1

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)

CTU1	ENST00000421832	NM_145232.3	ENSP00000390011	1