SNiPAcard

Block annotations

Block info

genomic range	chr16:57,442,191-57,444,002
block size	1,812 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.894 [-0.195 – 2.306]	**gene(s) hit or close-by**	CCL17
phastCons	μ = 0.060 [0.006 – 0.182]	**eQTL gene(s)**	–
GERP++	μ = 0.412 [-1.03 – 0.913]	**potentially regulated gene(s)**	–
CADD score	μ = 8.514 [5.131 – 13.24]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001633924 (CTCF binding site)	2	cervix (HeLa-S3)	CTCF, DNase1
		embryonic stem cell (H1ESC)	Rad21
		HSMMtube	DNase1, H3K27me3
		liver (HepG2)	Rad21, CTCF, H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	CTCF
ENSR00001505514 (enhancer)	2	embryonic stem cell (H1ESC)	Rad21
		HSMMtube	DNase1, H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	CTCF, DNase1
		liver (HepG2)	Rad21, CTCF, H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K4me1
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCL17	upstream gene variant	3779	ENST00000616880	?	ENSP00000480147	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCL17	ENST00000219244	NM_002987.2	ENSP00000219244	4