SNiPAcard

Block annotations

Block info

genomic range	chr1:248,022,712-248,027,609
block size	4,898 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.892 [-2.185 – 0.498]	**gene(s) hit or close-by**	TRIM58
phastCons	μ = 0.000 [0 – 0.001]	**eQTL gene(s)**	OR2T8, OR2W3, TRIM58
GERP++	μ = -1.524 [-7.78 – 1.54]	**potentially regulated gene(s)**	–
CADD score	μ = 2.793 [0.034 – 5.429]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
OR2W3	?	ENSG00000238243	tibial nerve	1.89×10^{-8} (p-value)	GTEx Portal V6	10
TRIM58	?	ENSG00000162722	tibial nerve	1.97×10^{-9} (p-value)	GTEx Portal V6	10
OR2T8	?	ENSG00000177462	tibial nerve	4.60×10^{-7} (p-value)	GTEx Portal V6	7
TRIM58	?	ENSG00000162722	blood	2.01×10^{-6} (p-value)	GTEx Portal V6	8
TRIM58	?	ENSG00000162722	muscularis mucosae	4.59×10^{-6} (p-value)	GTEx Portal V6	2
TRIM58	?	ENSG00000162722	unexposed skin	1.60×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000290792 (open chromatin region)	3	blood (K562)	DNase1, H3K9ac, H3K79me2, H3K4me3, H3K4me2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TRIM58	ENST00000366481	NM_015431.3	ENSP00000355437	D	gaT/gaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TRIM58	ENST00000366481	NM_015431.3	ENSP00000355437	9