Locus 298 (rs9651367)

SNiPAcard

Block annotations

Block info

genomic range	chr10:20,210,060-20,272,662
block size	62,603 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.225 [-1.003 – 0.583]	**gene(s) hit or close-by**	PLXDC2
phastCons	μ = 0.004 [0 – 0.027]	**eQTL gene(s)**	–
GERP++	μ = 0.007 [-3.75 – 2.28]	**potentially regulated gene(s)**	–
CADD score	μ = 4.352 [0.412 – 11.7]	**disease gene(s)**	–

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001421558 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1, H3K27ac, H3K9ac
		Osteobl	H3K4me2, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
		breast (HMEC)	DNase1, H3K27ac, H3K4me1, H3K4me2
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	FOXA1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K27ac
ENSR00000351639 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me2, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	H3K4me2, DNase1
ENSR00001594041 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H4K20me1
		endothelium (HUVEC)	Cjun, DNase1
		nervous (NH-A)	DNase1
ENSR00001594043 (enhancer)	1	monocytes (Monocytes-CD14+)	H4K20me1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLXDC2	ENST00000377242	NM_001282736.1	ENSP00000366450	9
PLXDC2	ENST00000377252	NM_032812.8	ENSP00000366460	9