SNiPAcard

Block annotations

Block info

genomic range	chr6:74,457,833-74,512,687
block size	54,855 bp
variant count	61 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.794 [-5.724 – 1.073]	**gene(s) hit or close-by**	CD109
phastCons	μ = 0.051 [0 – 1]	**eQTL gene(s)**	CD109
GERP++	μ = -0.676 [-5.57 – 5.16]	**potentially regulated gene(s)**	EEF1A1, RP11-554D15.1, RP11-9G10.1
CADD score	μ = 3.302 [0.038 – 15.22]	**disease gene(s)**	CD109

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Calcium levels	$<3.00\times10^{-6}$	GWAS Catalog	24068962	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM020385	1

Disease gene annotation

gene	trait	source DB	source entry/link
CD109	Fetal and neonatal alloimmune thrombocytopenia	OrphaNet	OrphaNet:853

Direct effect on transcript

Amino acid sequence alteration (splice region)

gene	effect type	affected transcript	RefSeq id	protein	amino acid	codon	SIFT prediction	PolyPhen prediction	varian(s)
CD109	missense variant	ENST00000287097	NM_133493.3	ENSP00000287097	Y/S	tAc/tCc	tolerated	benign	1
CD109	missense variant	ENST00000437994	NM_001159587.1	ENSP00000388062	Y/S	tAc/tCc	tolerated	benign	1
CD109	missense variant	ENST00000422508	NM_001159588.1	ENSP00000404475	Y/S	tAc/tCc	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD109	?	ENSG00000156535	tibial artery	6.50×10^{-8} (p-value)	GTEx Portal V6	60
CD109	?	ENSG00000156535	aorta	9.53×10^{-10} (p-value)	GTEx Portal V6	60

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000454297	1	ENCP00000049135	RP11-9G10.1
ENCE00000454282	2	ENCP00000049134	RP11-554D15.1
ENCE00000454216	1	ENCP00000049134	RP11-554D15.1
		ENCP00000049123	EEF1A1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001705222 (open chromatin region)	1	blood (K562)	H3K27me3
		A549	H3K36me3
ENSR00001705224 (enhancer)	2	skin (NHEK)	H3K36me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K36me3
ENSR00001220456 (promoter flanking region)	5	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K36me3, Rad21, CTCF
		HSMMtube	DNase1
		Osteobl	H3K27ac, CTCF
		skin (NHDF-AD)	H3K4me1, CTCF, DNase1
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF, DNase1
		cervix (HeLa-S3)	Cjun, Jund, CTCF
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	DNase1, CTCF
		A549	H3K36me3
		skin (NHEK)	CTCF
ENSR00001705225 (CTCF binding site)	4	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K36me3, Rad21, CTCF
		HSMMtube	DNase1
		Osteobl	CTCF, H3K27ac
		skin (NHDF-AD)	H3K4me1, CTCF, DNase1
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF, DNase1
		cervix (HeLa-S3)	Cjun, Jund, CTCF
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1, CTCF
		A549	H3K36me3
		nervous (NH-A)	CTCF, DNase1
		skin (NHEK)	CTCF
ENSR00001220457 (CTCF binding site)	1	skin (NHDF-AD)	H3K4me1
ENSR00001220458 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1, CTCF, H3K4me1
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	DNase1, CTCF
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF, DNase1
		Osteobl	H3K27ac, CTCF
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1, CTCF

	A549	H3K36me3, CTCF

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CD109	ENST00000422508	NM_001159588.1	ENSP00000404475	Y	taC/taT	1
CD109	ENST00000437994	NM_001159587.1	ENSP00000388062	Y	taC/taT	1
CD109	ENST00000287097	NM_133493.3	ENSP00000287097	Y	taC/taT	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CD109	ENST00000422508	NM_001159588.1	ENSP00000404475	59
CD109	ENST00000437994	NM_001159587.1	ENSP00000388062	59
CD109	ENST00000287097	NM_133493.3	ENSP00000287097	59
CD109	ENST00000474094	?	?	39