Locus 316 (rs11568819)

SNiPAcard

Block annotations

Block info

genomic range	chr11:102,397,005-102,401,633
block size	4,629 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.454 [-1.095 – 0.187]	gene(s) hit or close-by	MMP7
phastCons	μ = 0.348 [0 – 0.696]	eQTL gene(s)	MMP7
GERP++	μ = -2.955 [-6.39 – 0.48]	potentially regulated gene(s)	–
CADD score	μ = 6.064 [1.978 – 10.15]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR013972	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MMP7	?	ENSG00000137673	tibial nerve	3.83×10-7 (p-value)	GTEx Portal V6	2
MMP7	?	ENSG00000137673	thyroid	1.23×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000567974 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	H3K27me3
		A549	H3K4me2, H3K4me3
		nervous (NH-A)	DNase1
ENSR00001606494 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me2
		muscle (HSMM)	DNase1

cervix (HeLa-S3)	DNase1
endothelium (HUVEC)	Cjun, DNase1
liver (HepG2)	H3K27me3
A549	H3K4me2, H3K4me3
nervous (NH-A)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MMP7	downstream gene variant, upstream gene variant	155	ENST00000531200	?	?	2
MMP7	upstream gene variant	152	ENST00000533366	?	?	1
MMP7	upstream gene variant	149	ENST00000260227	NM_002423.3	ENSP00000260227	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MMP7	ENST00000533366	?	?	1
MMP7	ENST00000260227	NM_002423.3	ENSP00000260227	1