Locus 66 (rs35395347)

SNiPAcard

Block annotations

Block info

genomic range	chr3:49,277,998-49,335,745
block size	57,748 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.296 [-0.129 – 0.887]	gene(s) hit or close-by	CCDC36, USP4
phastCons	μ = 0.003 [0 – 0.013]	eQTL gene(s)	AMT, APEH, MRPS18AP1, MST1, NICN1, ZNF589
GERP++	μ = 0.732 [-1.39 – 1.88]	potentially regulated gene(s)	–
CADD score	μ = 3.251 [1.691 – 5.129]	disease gene(s)	MST1, AMT

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
MST1	Primary sclerosing cholangitis	OrphaNet	OrphaNet:171
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MRPS18AP1	?	ENSG00000229759	muscularis mucosae	1.57×10-6 (p-value)	GTEx Portal V6	1
MST1	?	ENSG00000173531	tibial nerve	4.13×10-5 (p-value)	GTEx Portal V6	5
MST1	?	ENSG00000173531	transformed fibroblasts	3.39×10-6 (p-value)	GTEx Portal V6	5
APEH	?	ENSG00000164062	transformed fibroblasts	1.99×10-5 (p-value)	GTEx Portal V6	5
MST1	?	ENSG00000173531	esophagus mucosa	2.20×10-6 (p-value)	GTEx Portal V6	5
MRPS18AP1	?	ENSG00000229759	esophagus mucosa	3.17×10-5 (p-value)	GTEx Portal V6	1
ZNF589	?	ENSG00000164048	esophagus mucosa	1.05×10-5 (p-value)	GTEx Portal V6	1
APEH	?	ENSG00000164062	esophagus mucosa	4.03×10-7 (p-value)	GTEx Portal V6	5
MST1	?	ENSG00000173531	subcutaneous adipocytes	2.11×10-6 (p-value)	GTEx Portal V6	5
NICN1	ENST00000273598	ILMN_1807211	blood	1.57×10-6 (p-value)	Westra et al.	1
NICN1	ENST00000615713					
AMT	ENST00000273588					
NICN1	ENST00000423832					
APEH	?	ENSG00000164062	unexposed skin	1.65×10-5 (p-value)	GTEx Portal V6	4
MST1	?	ENSG00000173531	thyroid	3.24×10-5 (p-value)	GTEx Portal V6	4

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001479015 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF
		HSMMtube	H3K36me3
		blood (K562)	Max, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF, H3K36me3
		liver (HepG2)	Rad21, CTCF, H3K36me3
		blood (GM12878)	Rad21, CTCF, H3K36me3
		lung (IMR90)	CTCF, H3K36me3
		nervous (NH-A)	CTCF, H3K36me3
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF, H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	CTCF, H3K36me3
		A549	H3K36me3, CTCF
ENSR00001479016 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K4me1, H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	H3K36me3, DNase1
		muscle (HSMM)	H3K27ac, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K27ac, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	DNase1, H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CCDC36	downstream gene variant	3991	ENST00000493870	?	?	1
USP4	upstream gene variant	1738	ENST00000475873	?	?	4
USP4	downstream gene variant	506	ENST00000486549	?	?	3
USP4	downstream gene variant	2283	ENST00000488520	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCDC36	ENST00000366429	?	ENSP00000403700	1
CCDC36	ENST00000296449	NM_178173.3	ENSP00000296449	1

CCDC36	ENST00000452691	NM_001135197.1	ENSP00000407837	1
CCDC36	ENST00000438782	?	ENSP00000391788	1
USP4	ENST00000485450	?	?	4
USP4	ENST00000265560	NM_003363.3	ENSP00000265560	4
USP4	ENST00000431357	?	ENSP00000399079	4
USP4	ENST00000351842	NM_199443.2	ENSP00000341028	4
USP4	ENST00000486549	?	?	1

Locus 66 (sentinel SNP rs35395347 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.