Locus 97 (rs41271951)

SNiPAcard

Block annotations

Block info

genomic range	chr1:150,616,888-150,737,220
block size	120,333 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.823 [-1.957 – -0.018]	gene(s) hit or close-by	CTSS, GOLPH3L, HORMAD1
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	CTSS, SEMA6C
GERP++	μ = -0.649 [-1.64 – 0.874]	potentially regulated gene(s)	CA14, CTSK
CADD score	μ = 4.745 [2.699 – 6.373]	disease gene(s)	CTSK

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CTSK	PYCNODYSOSTOSIS	OMIM	MIM:265800
CTSK	PYCNODYSOSTOSIS	DECIPHER	MIM:265800
CTSK	PYCNODYSOSTOSIS	OrphaNet	OrphaNet:763

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CTSS	missense variant	ENST00000368985	NM_004079.4	ENSP00000357981	A/V	gCg/gTg	?	?	1
CTSS	missense variant	ENST00000448301	NM_001199739.1	ENSP00000408414	A/V	gCg/gTg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CTSS	?	ENSG00000163131	muscularis mucosae	3.10×10^{-7} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	tibial nerve	2.48×10^{-12} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	transformed fibroblasts	7.49×10^{-10} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	tibial artery	2.61×10^{-13} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	thyroid	4.04×10^{-8} (p-value)	GTEx Portal V6	3
SEMA6C	?	ENSG00000143434	thyroid	2.60×10^{-8} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	skeletal muscle	6.75×10^{-9} (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	transverse colon	7.97×10^{-9} (p-value)	GTEx Portal V6	3
SEMA6C	?	ENSG00000143434	cerebellum	3.85×10^{-6} (p-value)	GTEx Portal V6	3

CTSS	?	ENSG00000163131	sun exposed skin	1.83×10-12 (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	subcutaneous adipocytes	5.77×10-13 (p-value)	GTEx Portal V6	3
CTSS	?	ENSG00000163131	pancreas	4.73×10-6 (p-value)	GTEx Portal V6	2
CTSS	?	ENSG00000163131	EBV lymphocytes	8.05×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000036221	1	ENCP00000004304	CA14
		ENCP00000004339	CTSK

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000544808 (promoter)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2
		blood (K562)	DNase1, PU1, H3K9ac, H2AZ, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	DNase1, Cjun
		liver (HepG2)	DNase1
		blood (GM12878)	H3K79me2, H3K4me3, H3K27ac, H3K9ac, DNase1, H2AZ, PU1, PolII
		lung (IMR90)	DNase1
		A549	H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K27ac, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTSS	upstream gene variant	42	ENST00000480760	?	?	1
GOLPH3L	downstream gene variant	4025	ENST00000427665	?	ENSP00000410476	1
GOLPH3L	downstream gene variant	1813	ENST00000271732	NM_018178.5	ENSP00000271732	1
HORMAD1	downstream gene variant	488	ENST00000476530	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HORMAD1	ENST00000322343	NM_001199829.1	ENSP00000326489	1
HORMAD1	ENST00000442853	?	ENSP00000401940	1
HORMAD1	ENST00000361824	NM_032132.4	ENSP00000355167	1
HORMAD1	ENST00000368987	?	ENSP00000357983	1
HORMAD1	ENST00000368995	?	ENSP00000357991	1