Locus 110 (rs6114248)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,692,121-23,731,560
block size	39,440 bp
variant count	41 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.414 [-4.432 – 0.242]	gene(s) hit or close-by	CST1, CST2P1
phastCons	μ = 0.013 [0 – 0.149]	eQTL gene(s)	CST1
GERP++	μ = -0.468 [-3.14 – 0.785]	potentially regulated gene(s)	CST11, CST8, CST9L, CSTL1, CSTP1, GGTLC1, XXyac-YX60D10.1
CADD score	μ = 1.308 [0.008 – 6.138]	disease gene(s)	–

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)
CST1	missense variant	ENST00000398402	?	ENSP00000381439	Y/H	Tat/Cat	deleterious	unknown	1
CST1	missense variant	ENST00000304749	NM_001898.2	ENSP00000305731	Y/H	Tat/Cat	deleterious	unknown	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST1	ENST00000398402	ILMN_1753449	skin	3.41×10-6 (p-value)	MuTHER consortium	9
CST1	ENST00000304749					
CST1	?	ENSG00000170373	testis	1.75×10-6 (p-value)	GTEx Portal V6	31

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290785	1	ENCP00000031084	CST9L
ENCE00000290706	1	ENCP00000031098	GGTLC1
		ENCP00000031079	CST11
		ENCP00000031078	CSTL1
		ENCP00000031093	XXyac-YX60D10.1
		ENCP00000031081	CST8
		ENCP00000031096	CSTP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000008781	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
(CTCF binding site)		HSMMtube	CTCF, H3K27me3, DNase1

		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, DNase1
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3, DNase1
		A549	CTCF, H3K27me3
ENSR00001664585 (enhancer)	2	embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000399221 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001448111 (CTCF binding site)	5	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST1	downstream gene variant	135	ENST00000398402	?	ENSP00000381439	4
CST1	downstream gene variant	33	ENST00000304749	NM_001898.2	ENSP00000305731	3
CST2P1	upstream gene variant	78	ENST00000425770	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST1	ENST00000304749	NM_001898.2	ENSP00000305731	9
CST1	ENST00000398402	?	ENSP00000381439	9

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST1	ENST00000304749	NM_001898.2	ENSP00000305731	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST1	ENST00000304749	NM_001898.2	ENSP00000305731	1