Locus 248 (rs7263120)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,673,017-23,727,735
block size	54,719 bp
variant count	53 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.786 [-4.406 – 0.696]	gene(s) hit or close-by	CST1, CST2P1, CST4
phastCons	μ = 0.015 [0 – 0.164]	eQTL gene(s)	CST1, CST3
GERP++	μ = -0.390 [-2.17 – 1.08]	potentially regulated gene(s)	CST11, CST4, CST8, CSTL1, CSTP1, GGTLC1, XXyac-YX60D10.1
CADD score	μ = 1.573 [0.012 – 7.042]	disease gene(s)	CST3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CST3	CEREBRAL AMYLOID ANGIOPATHY, CST3-RELATED	OMIM	MIM:105150
CST3	MACULAR DEGENERATION, AGE-RELATED, 11	OMIM	MIM:611953
CST3	Hereditary cerebral hemorrhage with amyloidosis, Icelandic type	OrphaNet	OrphaNet:100008

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST1	?	ENSG00000170373	tibial nerve	1.67×10^{-7} (p-value)	GTEx Portal V6	51
CST3	ENST00000398411	ILMN_1800354	adipocyte	2.52×10^{-5} (p-value)	MuTHER consortium	2
CST3	ENST00000376925					
CST3	ENST00000398409					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290703	1	ENCP00000031098	GGTLC1
		ENCP00000031079	CST11
		ENCP00000031078	CSTL1
		ENCP00000031093	XXyac-YX60D10.1
		ENCP00000031081	CST8
		ENCP00000031096	CSTP1
ENCE00000290742	1	ENCP00000031090	CST4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000399219 (promoter flanking region)	4	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	DNase1
		blood (DND-41)	H3K27me3

		skin (NHDF-AD)	DNase1, H3K4me1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	H3K27me3, DNase1
		lung (IMR90)	DNase1
ENSR00000399220 (enhancer)	2	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		liver (HepG2)	H3K4me1, H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST1	downstream gene variant	633	ENST00000398402	?	ENSP00000381439	8
CST1	downstream gene variant	455	ENST00000304749	NM_001898.2	ENSP00000305731	8
CST2P1	downstream gene variant, upstream gene variant	407	ENST00000425770	?	?	11
CST4	upstream gene variant	3340	ENST00000217423	NM_001899.2	ENSP00000217423	4