SNiPAcard

Block annotations

Block info

genomic range	chr20:23,810,857-23,848,024
block size	37,168 bp
variant count	37 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.968 [-3.14 – 0.428]	**gene(s) hit or close-by**	CST2
phastCons	μ = 0.017 [0 – 0.339]	**eQTL gene(s)**	CST1, CST2
GERP++	μ = -0.151 [-2.13 – 1.63]	**potentially regulated gene(s)**	RP4-564O4.1, RP5-1100I6.1
CADD score	μ = 1.878 [0.04 – 5.506]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST2	?	ENSG00000170369	lung	6.42×10^{-8} (p-value)	GTEx Portal V6	6
CST1	ENST00000398402	ILMN_1753449	blood	6.62×10^{-5} (p-value)	Westra et al.	10
CST1	ENST00000304749					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290875	1	ENCP00000031101	RP4-564O4.1
		ENCP00000031099	RP5-1100I6.1
ENCE00000290877	1	ENCP00000031101	RP4-564O4.1
		ENCP00000031099	RP5-1100I6.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001664592 (CTCF binding site)	2	HSMMtube	H3K27me3
ENSR00001664594 (TF binding site)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, USF1
		endothelium (HUVEC)	H3K36me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST2	upstream gene variant	3489	ENST00000304725	NM_001322.2	ENSP00000307540	3