Locus 90 (rs6138152)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,811,186-23,859,319
block size	48,134 bp
variant count	42 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.608 [-3.824 – 1.415]	gene(s) hit or close-by	CST2, CST5
phastCons	μ = 0.029 [0 – 0.353]	eQTL gene(s)	CST5
GERP++	μ = -0.237 [-3.17 – 1.19]	potentially regulated gene(s)	CST11, CST8, CSTL1, CSTP1, GGTLC1, RP4-564O4.1, RP5-1100I6.1
CADD score	μ = 2.157 [0.003 – 8.957]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Response to TNF antagonist treatment	$<3.00\times10^{-6}$	GWAS Catalog	18615156	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST5	?	ENSG00000170367	lung	1.25×10^{-9} (p-value)	GTEx Portal V6	42

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000290711	1	ENCP00000031098	GGTLC1
		ENCP00000031079	CST11
		ENCP00000031078	CSTL1
		ENCP00000031081	CST8
		ENCP00000031096	CSTP1
ENCE00000290878	1	ENCP00000031101	RP4-564O4.1
		ENCP00000031099	RP5-1100I6.1
ENCE00000290882	1	ENCP00000031101	RP4-564O4.1
		ENCP00000031099	RP5-1100I6.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001664593 (CTCF binding site)	1	HSMMtube	H3K27me3
ENSR00000233887 (open chromatin region)	2	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3

		A549	H3K27me3
ENSR00001664595 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF
		endothelium (HUVEC)	H3K36me3
		HSMMtube	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CST2	upstream gene variant	3818	ENST00000304725	NM_001322.2	ENSP00000307540	3
CST5	downstream gene variant	2403	ENST00000304710	NM_001900.4	ENSP00000307132	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CST5	ENST00000304710	NM_001900.4	ENSP00000307132	4