Locus 211 (rs2070856)

SNiPAcard

Block annotations

Block info

genomic range	chr20:23,731,412-23,731,412
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.296	gene(s) hit or close-by	CST1
phastCons	0	eQTL gene(s)	CST1
GERP++	-2.91	potentially regulated gene(s)	–
CADD score	0.003	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	L/P	cTg/cCg	?	?	1
CST1	missense variant	ENST00000304749	NM_001898.2	ENSP00000305731	L/P	cTg/cCg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CST1	?	ENSG00000170373	tibial nerve	2.38×10^{-6} (p-value)	GTEx Portal V6	1