

Block annotations

Block info

genomic range	chr20:23,731,412-23,731,412 <i>e!</i>
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.296	gene(s) hit or close-by	CST1 <i>e!</i>
phastCons	0	eQTL gene(s)	CST1 <i>e!</i>
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 <i>e!</i>	missense variant	ENST00000398402 <i>e!</i>	?	ENSP00000381439 L/P		cTg/cCg	?	?	1
CST1 <i>e!</i>	missense variant	ENST00000304749 <i>e!</i>	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 <i>e!</i>	?	ENSG00000170373 <i>e!</i>	tibial nerve	2.38×10 ⁻⁶ (p-value)	GTEx Portal V6 <i>IM</i>	1

