SNiPAcard

Block annotations

Block info

genomic range	chr1:57,422,484-57,422,484
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.535	**gene(s) hit or close-by**	C8B
phastCons	0.998	**eQTL gene(s)**	–
GERP++	-0.165	**potentially regulated gene(s)**	–
CADD score	17.12	**disease gene(s)**	C8B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
C8B	Immunodeficiency due to a late component of complements deficiency	OrphaNet	OrphaNet:169150	orphanet

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)
C8B	missense variant	ENST00000535057	NM_001278544.1	ENSP00000440113	G/R	Gga/Aga	tolerated	benign	1
C8B	missense variant	ENST00000543257	NM_001278543.1	ENSP00000442548	G/R	Gga/Aga	tolerated	benign	1
C8B	missense variant	ENST00000371237	NM_000066.3	ENSP00000360281	G/R	Gga/Aga	tolerated	benign	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C8B	downstream gene variant	44	ENST00000468990	?	?	1
C8B	downstream gene variant	3252	ENST00000494324	?	?	1