SNiPAcard

Block annotations

Block info

genomic range	chr1:57,412,848-57,428,589
block size	15,742 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.766 [-1.388 – 0.056]	**gene(s) hit or close-by**	AL161740.1, C8B, RP5-1103B4.3
phastCons	μ = 0.001 [0 – 0.003]	**eQTL gene(s)**	–
GERP++	μ = -2.208 [-4.95 – 1.85]	**potentially regulated gene(s)**	–
CADD score	μ = 2.995 [0.285 – 6.988]	**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

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL161740.1	upstream gene variant	3364	ENST00000408664	?	?	1
C8B	downstream gene variant	2805	ENST00000494324	?	?	1
RP5-1103B4.3	upstream gene variant	970	ENST00000417420	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C8B	ENST00000468990	?	?	3
C8B	ENST00000543257	NM_001278543.1	ENSP00000442548	6
C8B	ENST00000535057	NM_001278544.1	ENSP00000440113	6
C8B	ENST00000371237	NM_000066.3	ENSP00000360281	6
C8B	ENST00000494324	?	?	2