Locus 173 (rs2786107)

SNiPAcard

Block annotations

Block info

genomic range	chr1:197,310,777-197,310,777
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.427	gene(s) hit or close-by	CRB1
phastCons	0	eQTL gene(s)	–
GERP++	-1.17	potentially regulated gene(s)	–
CADD score	1.07	disease gene(s)	CRB1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CRB1	LEBER CONGENITAL AMAUROSIS 8	DECIPHER	MIM:613835
CRB1	Retinitis pigmentosa-12, autosomal recessive	DECIPHER	MIM:600105
CRB1	RETINITIS PIGMENTOSA 12	OMIM	MIM:600105
CRB1	LEBER CONGENITAL AMAUROSIS 8	OMIM	MIM:613835
CRB1	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY	OMIM	MIM:172870
CRB1	RETINITIS PIGMENTOSA	OMIM	MIM:268000
CRB1	Retinitis pigmentosa	OrphaNet	OrphaNet:791
CRB1	Pigmented paravenous retinochoroidal atrophy	OrphaNet	OrphaNet:251295
CRB1	Leber congenital amaurosis	OrphaNet	OrphaNet:65

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CRB1	ENST00000367399	NM_001193640.1	ENSP00000356369	1
CRB1	ENST00000475659	?	?	1
CRB1	ENST00000484075	?	ENSP00000433932	1
CRB1	ENST00000538660	NM_001257966.1	ENSP00000438091	1
CRB1	ENST00000535699	NM_001257965.1	ENSP00000438786	1
CRB1	ENST00000367400	NM_201253.2	ENSP00000356370	1