Locus 112 (rs419137)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,696,875-196,696,875
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-1.535	gene(s) hit or close-by	CFH
phastCons	0	eQTL gene(s)	–
GERP++	-4.04	potentially regulated gene(s)	–
CADD score	0.742	disease gene(s)	CFH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Macular degeneration	2.33×10^{-9}	dbGaP	pha002890	dbGaP	1
Macular degeneration	1.06×10^{-10}	dbGaP	pha002869	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421	orphanet
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579	orphanet
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571	orphanet
CFH	Familial drusen	OrphaNet	OrphaNet:75376	orphanet
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903	orphanet

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000466229	?	?	1
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	1