SNiPAcard

Block annotations

Block info

genomic range	chr1:196,675,239-196,692,940
block size	17,702 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.300 [-2.22 – -0.379]	**gene(s) hit or close-by**	CFH
phastCons	μ = 0.004 [0 – 0.007]	**eQTL gene(s)**	–
GERP++	μ = -1.285 [-1.44 – -1.13]	**potentially regulated gene(s)**	–
CADD score	μ = 0.389 [0.055 – 0.723]	**disease gene(s)**	CFH

Trait annotations

Disease gene annotation

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

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000550120 (CTCF binding site)	1	NHLF	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	DNase1
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K36me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF, H3K36me3
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF
		A549	CTCF
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFH	downstream gene variant	4544	ENST00000359637	?	ENSP00000352658	1
CFH	downstream gene variant	4543	ENST00000630130	NM_001014975.2	ENSP00000487250	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000466229	?	?	2
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	2