SNiPAcard

Block annotations

Block info

genomic range	chr1:196,675,861-196,675,861
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.57	**gene(s) hit or close-by**	CFH
phastCons	0.001	**eQTL gene(s)**	CFHR3
GERP++	0.628	**potentially regulated gene(s)**	–
CADD score	2.058	**disease gene(s)**	CFHR3, CFH

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	atrial appendage	7.18×10^{-6} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	tibial nerve	7.49×10^{-10} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	aorta	6.75×10^{-7} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	tibial artery	3.96×10^{-9} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	subcutaneous adipocytes	2.39×10^{-7} (p-value)	GTEx Portal V6	1

Putative effect on transcript

Intron variant

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