Locus 76 (rs10801582)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,845,273-196,944,357
block size	99,085 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.006 [-1.277 – 0.754]	**gene(s) hit or close-by**	CFHR2, CFHR2, CFHR4, CFHR5, RP4-608O15.4
phastCons	μ = 0.095 [0 – 0.492]	**eQTL gene(s)**	CFH, CFHR1, CFHR3, CFHR4
GERP++	μ = -0.192 [-3.63 – 1.43]	**potentially regulated gene(s)**	–
CADD score	μ = 4.973 [0.282 – 13.66]	**disease gene(s)**	CFHR1, CFHR3, CFH, CFHR5

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
End-stage coagulation	$<7.00\times10^{-19}$	GWAS Catalog	23381943	1
Macular degeneration	5.49×10^{-30}	dbGaP	pha002890	1
Macular degeneration	5.91×10^{-34}	dbGaP	pha002869	1
Macular degeneration	6.21×10^{-5}	dbGaP	pha002856	1
Macular degeneration	2.51×10^{-6}	dbGaP	pha000001	2

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR1	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFHR1	C3 glomerulonephritis	OrphaNet	OrphaNet:329931
CFHR1	Dense deposit disease	OrphaNet	OrphaNet:93571
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421
CFH	Dense deposit disease	OrphaNet	OrphaNet:93571
CFH	Familial drusen	OrphaNet	OrphaNet:75376
CFH	Immunoglobulin-mediated membranoproliferative glomerulonephritis	OrphaNet	OrphaNet:329903
CFHR5	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581
CFHR5	C3 glomerulonephritis	OrphaNet	OrphaNet:329931
CFHR5	CFHR5 DEFICIENCY	OMIM	MIM:614809

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	lung	4.99×10^{-6} (p-value)	GTEx Portal V6	1
CFHR1	?	ENSG00000244414	tibial nerve	1.40×10^{-6} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	tibial nerve	9.52×10^{-11} (p-value)	GTEx Portal V6	12
CFH	?	ENSG00000000971	tibial nerve	2.60×10^{-6} (p-value)	GTEx Portal V6	13

CFHR4	?	ENSG00000134365	liver	6.84×10-8 (p-value)	GTEx Portal V6	16
CFHR1	?	ENSG00000244414	subcutaneous adipocytes	1.65×10-8 (p-value)	GTEx Portal V6	16
CFHR3	?	ENSG00000116785	subcutaneous adipocytes	7.04×10-11 (p-value)	GTEx Portal V6	16
CFHR3	?	ENSG00000116785	thyroid	4.81×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFHR2	downstream gene variant	965	ENST00000473386	?	?	1
CFHR2	downstream gene variant	970	ENST00000367421	?	ENSP00000356391	1
CFHR2	downstream gene variant	954	ENST00000476712	?	ENSP00000476677	1
CFHR2	downstream gene variant	79	ENST00000489703	?	?	3
CFHR2	downstream gene variant	954	ENST00000367415	NM_005666.2	ENSP00000356385	1
CFHR2	downstream gene variant	142	ENST00000485647	?	?	2
CFHR2	downstream gene variant	961	ENST00000496448	?	?	1
CFHR4	upstream gene variant	1523	ENST00000251424	NM_006684.4	ENSP00000251424	1
CFHR4	upstream gene variant	1455	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	1
CFHR4	upstream gene variant	1544	ENST00000367418	?	ENSP00000356388	1
CFHR5	upstream gene variant	2329	ENST00000256785	NM_030787.3	ENSP00000256785	1
RP4-608O15.4	downstream gene variant	4556	ENST00000613613	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CFHR2	ENST00000367421	?	ENSP00000356391	C	tgT/tgC	1
CFHR2	ENST00000367415	NM_005666.2	ENSP00000356385	C	tgT/tgC	1
CFHR2	ENST00000476712	?	ENSP00000476677	C	tgT/tgC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFHR2	ENST00000473386	?	?	4
CFHR2	ENST00000367421	?	ENSP00000356391	16
CFHR2	ENST00000485647	?	?	2
CFHR2	ENST00000367415	NM_005666.2	ENSP00000356385	4
CFHR2	ENST00000489703	?	?	1
CFHR2	ENST00000476712	?	ENSP00000476677	4
CFHR2	ENST00000496448	?	?	4
CFHR4	ENST00000251424	NM_006684.4	ENSP00000251424	6
CFHR4	ENST00000608469	?	ENSP00000477162	8

CFHR4	ENST00000367418	?	ENSP00000356388	6
CFHR4	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	6

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CFHR2	ENST00000485647	?	1
CFHR2	ENST00000496448	?	1