Locus 259 (rs12734260)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,719,947-196,937,951
block size	218,005 bp
variant count	70 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.211 [-2.649 – 2.266]	**gene(s) hit or close-by**	CFH, CFHR2, CFHR2, CFHR4, RP4-608O15.4
phastCons	μ = 0.069 [0 – 0.987]	**eQTL gene(s)**	CFH, CFHR1, CFHR3, JUP, ZBTB41
GERP++	μ = -0.271 [-2.54 – 1.61]	**potentially regulated gene(s)**	KCNT2
CADD score	μ = 2.963 [0.062 – 14.82]	**disease gene(s)**	CFHR1, CFHR3, CFH, JUP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
HWESASXX*	1.65×10^{-5}	Metabolomics GWAS Server	24816252	8
Macular degeneration	5.99×10^{-20}	dbGaP	pha002890	5
Macular degeneration	4.93×10^{-21}	dbGaP	pha002869	3
Macular degeneration	4.26×10^{-11}	dbGaP	pha000002	1

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	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
JUP	Lethal acantholytic epidermolysis bullosa	OrphaNet	OrphaNet:158687
JUP	NAXOS DISEASE	OrphaNet	OrphaNet:34217
JUP	Familial isolated arrhythmogenic ventricular dysplasia, right dominant form	OrphaNet	OrphaNet:293910
JUP	Familial isolated arrhythmogenic ventricular dysplasia, left dominant form	OrphaNet	OrphaNet:293888
JUP	Familial isolated arrhythmogenic ventricular dysplasia, biventricular form	OrphaNet	OrphaNet:293899
JUP	NAXOS DISEASE	OMIM	MIM:601214
JUP	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 12	OMIM	MIM:611528

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	muscularis mucosae	1.30×10^{-8} (p-value)	GTEx Portal V6	68
CFHR3	?	ENSG00000116785	adrenal gland	5.83×10^{-10} (p-value)	GTEx Portal V6	66

CFHR3	?	ENSG00000116785	lung	1.03×10-13 (p-value)	GTEx Portal V6	68
CFHR1	?	ENSG00000244414	transformed fibroblasts	9.59×10-7 (p-value)	GTEx Portal V6	58
CFHR1	?	ENSG00000244414	liver	3.11×10-9 (p-value)	GTEx Portal V6	67
CFHR3	?	ENSG00000116785	liver	2.62×10-7 (p-value)	GTEx Portal V6	60
CFHR3	?	ENSG00000116785	thyroid	4.09×10-12 (p-value)	GTEx Portal V6	68
CFHR3	?	ENSG00000116785	prostate	3.24×10-6 (p-value)	GTEx Portal V6	4
CFHR3	?	ENSG00000116785	transverse colon	7.15×10-7 (p-value)	GTEx Portal V6	51
CFHR3	?	ENSG00000116785	tibial nerve	3.80×10-6 (p-value)	GTEx Portal V6	46
CFHR3	?	ENSG00000116785	esophagus mucosa	1.67×10-6 (p-value)	GTEx Portal V6	47
CFHR3	?	ENSG00000116785	aorta	4.24×10-8 (p-value)	GTEx Portal V6	53
CFH	ENST00000359637	ILMN_1657803	skin	4.41×10-7 (p-value)	MuTHER consortium	23
CFH	ENST00000630130		adipocyte	3.96×10-7 (p-value)	MuTHER consortium	23
CFHR3	?	ENSG00000116785	stomach	3.95×10-6 (p-value)	GTEx Portal V6	10
ZBTB41	?	ENSG00000177888	subcutaneous adipocytes	2.08×10-5 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
JUP	?	ENSG00000173801	chr17	blood	4.64×10-2 (q-value)	SeeQTL DB (HapMap)	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000048799	1	ENCP00000005933	KCNT2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001527235 (TF binding site)	1	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	H3K27me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFH	downstream gene variant	3313	ENST00000466229	?	?	1
CFH	downstream gene variant	3313	ENST00000367429	NM_000186.3	ENSP00000356399	1
CFHR2	upstream gene variant, downstream gene variant	745	ENST00000473386	?	?	4
CFHR2	upstream gene variant, downstream gene variant	714	ENST00000496448	?	?	4
CFHR2	upstream gene variant, downstream gene variant	659	ENST00000485647	?	?	5
CFHR2	downstream gene variant	4905	ENST00000367421	?	ENSP00000356391	1

CFHR2	upstream gene variant, downstream gene variant	668	ENST00000367415	NM_005666.2	ENSP00000356385	4
CFHR2	upstream gene variant, downstream gene variant	659	ENST00000476712	?	ENSP00000476677	4
CFHR2	upstream gene variant, downstream gene variant	704	ENST00000489703	?	?	4
CFHR4	upstream gene variant, downstream gene variant	3978	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	2
CFHR4	downstream gene variant	4220	ENST00000608469	?	ENSP00000477162	1
CFHR4	upstream gene variant, downstream gene variant	4046	ENST00000251424	NM_006684.4	ENSP00000251424	2
CFHR4	upstream gene variant, downstream gene variant	4067	ENST00000367418	?	ENSP00000356388	2
RP4-608O15.4	upstream gene variant	1674	ENST00000613613	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFHR2	ENST00000473386	?	?	4
CFHR2	ENST00000367421	?	ENSP00000356391	65
CFHR2	ENST00000485647	?	?	2
CFHR2	ENST00000476712	?	ENSP00000476677	4
CFHR2	ENST00000489703	?	?	2
CFHR2	ENST00000367415	NM_005666.2	ENSP00000356385	4
CFHR2	ENST00000496448	?	?	4
CFHR4	ENST00000251424	NM_006684.4	ENSP00000251424	12
CFHR4	ENST00000608469	?	ENSP00000477162	51
CFHR4	ENST00000367418	?	ENSP00000356388	12
CFHR4	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	12