SNiPAcard

Block annotations

Block info

genomic range	chr1:196,646,176-196,717,153
block size	70,978 bp
variant count	77 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.104 [-7.227 – 2.829]	**gene(s) hit or close-by**	CFH
phastCons	μ = 0.040 [0 – 0.596]	**eQTL gene(s)**	CFH, CFHR1, CFHR3, RP11-75C23.1
GERP++	μ = -0.621 [-9.79 – 2.77]	**potentially regulated gene(s)**	–
CADD score	μ = 1.853 [0.001 – 8.248]	**disease gene(s)**	CFHR1, CFHR3, CFH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Age-related macular degeneration	$<1.00\times10^{-31}$	GWAS Catalog	23577725	1
Age-related macular degeneration (extreme sampling)	$<9.00\times10^{-24}$	GWAS Catalog	23577725	1
Age-related macular degeneration	$<7.00\times10^{-32}$	GWAS Catalog	23326517	2
Age-related macular degeneration (CNV)	$<1.00\times10^{-108}$	GWAS Catalog	22705344	1
Age-related macular degeneration (GA)	$<5.00\times10^{-67}$	GWAS Catalog	22705344	1
Age-related macular degeneration	$<2.00\times10^{-47}$	GWAS Catalog	22694956	1
Age-related macular degeneration	$<1.00\times10^{-261}$	GWAS Catalog	21665990	1
Age-related macular degeneration	$<4.00\times10^{-117}$	GWAS Catalog	20385826	1
Age-related macular degeneration	$<6.00\times10^{-16}$	GWAS Catalog	20385819	1
Age-related macular degeneration	$<4.00\times10^{-8}$	GWAS Catalog	15761122	1
HWESASXX*	2.58×10^{-6}	Metabolomics GWAS Server	24816252	4
Macular degeneration	9.52×10^{-75}	dbGaP	pha002890	3
Macular degeneration	2.07×10^{-88}	dbGaP	pha002869	3
Macular degeneration	6.04×10^{-18}	dbGaP	pha002856	4
Macular degeneration	1.75×10^{-19}	dbGaP	pha000002	1
Macular degeneration	1.29×10^{-25}	dbGaP	pha000001	4

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Basal laminar drusen	pathogenic	ClinVar	RCV000018016.27	1
Age-related macular degeneration 4	risk factor	ClinVar	RCV000018015.2	1
?	HGMD curated	HGMD	CM057396	1
?	HGMD curated	HGMD	CM051020	1
MACULAR DEGENERATION, AGE-RELATED, 4, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:134370	1

Disease gene annotation

gene	trait	source DB	source entry/link

CFHR1	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFHR1	C3 glomerulonephritis	OrphaNet	OrphaNet:329931	orphanet
CFHR1	Dense deposit disease	OrphaNet	OrphaNet:93571	orphanet
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFH	Atypical hemolytic uremic syndrome with H factor anomaly	OrphaNet	OrphaNet:93579	orphanet
CFH	Immunodeficiency with factor H anomaly	OrphaNet	OrphaNet:200421	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

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CFH	missense variant	ENST00000359637	?	ENSP00000352658	H/Y	Cat/Tat	tolerated	benign	1
CFH	missense variant	ENST00000630130	NM_001014975.2	ENSP00000487250	H/Y	Cat/Tat	tolerated	benign	1
CFH	missense variant	ENST00000367429	NM_000186.3	ENSP00000356399	H/Y	Cat/Tat	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFH	ENST00000359637	ILMN_1657803	skin	9.43×10^{-8} (p-value)	MuTHER consortium	33
CFH	ENST00000630130		adipocyte	1.02×10^{-5} (p-value)	MuTHER consortium	33
CFH	ENST00000466229	ILMN_1810910	blood	4.35×10^{-6} (p-value)	Westra et al.	32
CFH	ENST00000359637					
CFH	ENST00000367429					
CFH	ENST00000630130					
CFHR1	?	ENSG00000244414	liver	6.46×10^{-8} (p-value)	GTEx Portal V6	75
CFHR3	?	ENSG00000116785	thyroid	5.91×10^{-7} (p-value)	GTEx Portal V6	26
CFH	?	ENSG00000000971	tibial nerve	2.39×10^{-7} (p-value)	GTEx Portal V6	36
CFHR1	?	ENSG00000244414	subcutaneous adipocytes	4.65×10^{-8} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	subcutaneous adipocytes	2.27×10^{-8} (p-value)	GTEx Portal V6	1
RP11-75C23.1	?	ENSG00000230260	nucleus accumbens	6.02×10^{-6} (p-value)	GTEx Portal V6	5
CFHR1	?	ENSG00000244414	transformed fibroblasts	3.21×10^{-5} (p-value)	GTEx Portal V6	5
CFHR3	?	ENSG00000116785	adrenal gland	6.26×10^{-6} (p-value)	GTEx Portal V6	4
CFHR3	?	ENSG00000116785	muscularis mucosae	3.73×10^{-6} (p-value)	GTEx Portal V6	3
CFHR1	?	ENSG00000244414	spleen	5.75×10^{-6} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	transverse colon	1.31×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	16	no	0.004	1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000550120 (CTCF binding site)	3	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	3639	ENST00000496761	?	?	2
CFH	downstream gene variant	519	ENST00000466229	?	?	1
CFH	downstream gene variant	62	ENST00000359637	?	ENSP00000352658	15
CFH	upstream gene variant	112	ENST00000470918	?	?	4
CFH	downstream gene variant	61	ENST00000630130	NM_001014975.2	ENSP00000487250	15
CFH	downstream gene variant	519	ENST00000367429	NM_000186.3	ENSP00000356399	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CFH	ENST00000630130	NM_001014975.2	ENSP00000487250	A	gcA/gcC	1
CFH	ENST00000359637	?	ENSP00000352658	A	gcA/gcC	1
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	A	gcA/gcC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000630130	NM_001014975.2	ENSP00000487250	36
CFH	ENST00000466229	?	?	74
CFH	ENST00000359637	?	ENSP00000352658	36
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	74

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CFH	ENST00000466229	?	4