SNiPAcard

Block annotations

Block info

genomic range	chr1:196,679,455-196,704,632
block size	25,178 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.440 [-1.946 – 0.687]	**gene(s) hit or close-by**	CFH
phastCons	μ = 0.024 [0 – 0.272]	**eQTL gene(s)**	CFH, CFHR1, CFHR3
GERP++	μ = -1.327 [-6.86 – 1.73]	**potentially regulated gene(s)**	–
CADD score	μ = 1.228 [0.015 – 5.622]	**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	$<1.00\times10^{-434}$	GWAS Catalog	23455636	1
End-stage coagulation	$<1.00\times10^{-9}$	GWAS Catalog	23381943	1
Age-related macular degeneration	$<3.00\times10^{-64}$	GWAS Catalog	20861866	1
Age-related macular degeneration	$<2.00\times10^{-111}$	GWAS Catalog	20385826	1
Age-related macular degeneration	$<2.00\times10^{-76}$	GWAS Catalog	20385819	1
HWESASXX*	2.20×10^{-8}	Metabolomics GWAS Server	24816252	1
Macular degeneration	6.93×10^{-76}	dbGaP	pha002890	1
Macular degeneration	8.92×10^{-92}	dbGaP	pha002869	1
Macular degeneration	1.35×10^{-16}	dbGaP	pha000002	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Age-related macular degeneration 4	risk factor	ClinVar	RCV000018024.1	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
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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFH	ENST00000359637	ILMN_1657803	skin	2.03×10^{-7} (p-value)	MuTHER consortium	6
CFH	ENST00000630130		adipocyte	3.54×10^{-7} (p-value)	MuTHER consortium	6
CFHR1	?	ENSG00000244414	transformed fibroblasts	5.58×10^{-6} (p-value)	GTEx Portal V6	19
CFHR1	?	ENSG00000244414	liver	4.07×10^{-10} (p-value)	GTEx Portal V6	19
CFHR3	?	ENSG00000116785	liver	8.03×10^{-7} (p-value)	GTEx Portal V6	19
CFHR1	?	ENSG00000244414	subcutaneous adipocytes	1.52×10^{-6} (p-value)	GTEx Portal V6	19
CFH	?	ENSG00000000971	subcutaneous adipocytes	6.47×10^{-6} (p-value)	GTEx Portal V6	19

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CFH	upstream gene variant	1406	ENST00000470918	?	?	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFH	ENST00000466229	?	?	19
CFH	ENST00000367429	NM_000186.3	ENSP00000356399	19