Locus 301 (rs6003)

SNiPAcard

Block annotations

Block info

genomic range	chr1:196,945,050-197,257,556
block size	312,507 bp
variant count	155 variants

Basic features

Conservation/deleteriousness			Linked genes
phyloP	μ = -0.701 [-7.024 – 2.567]	gene(s) hit or close-by	ASPM, CFHR5, CRB1, F13B, RP11-32D17.4, RP11-332L8.1, ZBTB41
phastCons	μ = 0.052 [0 – 1]	eQTL gene(s)	CFHR1, CFHR3, ZBTB41
GERP++	μ = -0.725 [-9.06 – 3.26]	potentially regulated gene(s)	ASPM, CFH, CFH, CFHR1, CFHR1, CFHR2, CFHR2, CFHR3, CFHR3, CFHR4
CADD score	μ = 3.100 [0.004 – 17.66]	disease gene(s)	CFHR5, F13B, ASPM, CRB1, CFHR3, CFHR1, CFH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Macular degeneration	6.24×10^{-11}	dbGaP	pha002890	2
Macular degeneration	7.66×10^{-12}	dbGaP	pha002869	2
Macular degeneration	4.75×10^{-6}	dbGaP	pha002856	9
Macular degeneration	4.57×10^{-9}	dbGaP	pha000002	3
Macular degeneration	2.17×10^{-9}	dbGaP	pha000001	9

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000145187.5	1
Clinvar: phenotype not specified	likely benign	ClinVar	RCV000145113.1	1
Clinvar: phenotype not specified	likely benign	ClinVar	RCV000145090.1	1
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000145076.2	1
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000081938.6	1
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000081936.6	1
Primary autosomal recessive microcephaly 5	benign	ClinVar	RCV000020792.1	1
Primary autosomal recessive microcephaly 5	benign	ClinVar	RCV000020768.1	1
Venous thrombosis, susceptibility to	risk factor	ClinVar	RCV000017984.1	1
?	HGMD curated	HGMD	CR063400	1
?	HGMD curated	HGMD	CR063399	1
?	HGMD curated	HGMD	CM034655	1
VENOUS THROMBOSIS, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:134580	1

Disease gene annotation

gene	trait	source DB	source entry/link
CFHR5	CFHR5 DEFICIENCY	OMIM	MIM:614809
F13B	FACTOR XIII, B SUBUNIT, DEFICIENCY OF	OMIM	MIM:613235

ASPM	MICROCEPHALY 5, PRIMARY, AUTOSOMAL RECESSIVE	OMIM	MIM:608716	OMIM
CRB1	RETINITIS PIGMENTOSA 12	OMIM	MIM:600105	OMIM
CRB1	LEBER CONGENITAL AMAUROSIS 8	OMIM	MIM:613835	OMIM
CRB1	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY	OMIM	MIM:172870	OMIM
CRB1	RETINITIS PIGMENTOSA	OMIM	MIM:268000	OMIM
CFHR3	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFHR5	Atypical hemolytic-uremic syndrome with anti-factor H antibodies	OrphaNet	OrphaNet:93581	orphanet
CFHR5	C3 glomerulonephritis	OrphaNet	OrphaNet:329931	orphanet
F13B	Congenital factor XIII deficiency	OrphaNet	OrphaNet:331	orphanet
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
ASPM	Autosomal recessive primary microcephaly	OrphaNet	OrphaNet:2512	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
CRB1	Retinitis pigmentosa	OrphaNet	OrphaNet:791	orphanet
CRB1	Pigmented paravenous retinochoroidal atrophy	OrphaNet	OrphaNet:251295	orphanet
CRB1	Leber congenital amaurosis	OrphaNet	OrphaNet:65	orphanet
ASPM	Primary Autosomal Recessive Microcephaly	DECIPHER	MIM:608716	OMIM
CRB1	LEBER CONGENITAL AMAUROSIS 8	DECIPHER	MIM:613835	OMIM
CRB1	Retinitis pigmentosa-12, autosomal recessive	DECIPHER	MIM:600105	OMIM

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)
F13B	missense variant	ENST00000367412	NM_001994.2	ENSP00000356382	H/R	cAt/cGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CFHR3	?	ENSG00000116785	lung	1.59×10^{-9} (p-value)	GTEx Portal V6	133
CFHR3	?	ENSG00000116785	aorta	1.12×10^{-6} (p-value)	GTEx Portal V6	21
ZBTB41	?	ENSG00000177888	subcutaneous adipocytes	8.07×10^{-8} (p-value)	GTEx Portal V6	127
CFHR3	?	ENSG00000116785	thyroid	4.38×10^{-10} (p-value)	GTEx Portal V6	130
CFHR3	?	ENSG00000116785	prostate	3.26×10^{-6} (p-value)	GTEx Portal V6	1
CFHR1	?	ENSG00000244414	transformed fibroblasts	2.75×10^{-5} (p-value)	GTEx Portal V6	1
CFHR3	?	ENSG00000116785	muscularis mucosae	1.98×10^{-5} (p-value)	GTEx Portal V6	2
CFHR3	?	ENSG00000116785	adrenal gland	1.18×10^{-5} (p-value)	GTEx Portal V6	4

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000048862	1	ENCP00000005943	CFHR2 CFHR2
		ENCP00000005941	CFHR1 CFHR1
ENCE00000048832	1	ENCP00000005947	ASPM
		ENCP00000005938	CFH CFH
ENCE00000048835	1	ENCP00000005942	CFHR4
		ENCP00000005939	CFHR3 CFHR3

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001527237 (CTCF binding site)	1	skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001589240 (enhancer)	1	skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00001589241 (CTCF binding site)	1	liver (HepG2)	CTCF
ENSR00001527239 (promoter flanking region)	1	HSMMtube	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000288050 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	DNase1
		A549	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000550137 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00001527244 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K36me3
ENSR00001589251 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001527245 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001589253 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3

		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001527246 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H2AZ
		blood (K562)	H3K79me2, H3K36me3
		blood (DND-41)	H3K4me2, H3K36me3, H3K4me1
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (GM12878)	H2AZ, H3K79me2, H3K4me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
ENSR00000550147 (promoter flanking region)	4	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (K562)	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K4me1, HNF4A, FOXA1, H3K27ac
		blood (GM12878)	H3K36me3
		A549	DNase1, H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000550149 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K27ac, CTCF, H3K36me3
		blood (K562)	DNase1, CTCF, H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	DNase1, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		A549	DNase1, H3K27ac, H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001527247 (promoter flanking region)	1	cervix (HeLa-S3)	H3K4me2
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (GM12878)	H3K4me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K4me2
		muscle (HSMM)	H3K36me3
ENSR00001589259 (enhancer)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ASPM	1	ENST00000294732 ENST00000367408 ENST00000367409	hsa-miR-382-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ASPM	downstream gene variant, upstream gene variant	454	ENST00000294732	NM_001206846.1	ENSP00000294732	6
ASPM	downstream gene variant, upstream gene variant	458	ENST00000367408	?	ENSP00000356378	11
ASPM	downstream gene variant, upstream gene variant	930	ENST00000612785	?	ENSP00000479244	5
ASPM	downstream gene variant, upstream gene variant	454	ENST00000367409	NM_018136.4	ENSP00000356379	6
CFHR5	upstream gene variant, downstream gene variant	140	ENST00000256785	NM_030787.3	ENSP00000256785	9
F13B	downstream gene variant, upstream gene variant	471	ENST00000367412	NM_001994.2	ENSP00000356382	11
F13B	downstream gene variant, upstream gene variant	1471	ENST00000490002	?	?	7
RP11-32D17.4	downstream gene variant, upstream gene variant	256	ENST00000442280	?	?	10
RP11-332L8.1	upstream gene variant	2444	ENST00000417716	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ASPM	ENST00000612785	?	ENSP00000479244	L	ttG/ttA	3
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	1	2	3
ASPM	ENST00000367408	?	ENSP00000356378	S	tcA/tcT	2
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	2	3	3
F13B	ENST00000367412	NM_001994.2	ENSP00000356382	T	acG/acA	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	1
ASPM	ENST00000367408	?	ENSP00000356378	1
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ASPM	ENST00000612785	?	ENSP00000479244	45
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	46
ASPM	ENST00000367408	?	ENSP00000356378	35
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	46
CFHR5	ENST00000256785	NM_030787.3	ENSP00000256785	11
CRB1	ENST00000475659	?	?	4
CRB1	ENST00000538660	NM_001257966.1	ENSP00000438091	4
CRB1	ENST00000535699	NM_001257965.1	ENSP00000438786	4
CRB1	ENST00000484075	?	ENSP00000433932	4
CRB1	ENST00000367400	NM_201253.2	ENSP00000356370	4
CRB1	ENST00000367399	NM_001193640.1	ENSP00000356369	4
F13B	ENST00000490002	?	?	7
F13B	ENST00000367412	NM_001994.2	ENSP00000356382	17
ZBTB41	ENST00000367405	NM_194314.2	ENSP00000356375	24

ZBTB41	ENST00000467322	?	?	24

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	1
ASPM	ENST00000367408	?	ENSP00000356378	1
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CFHR5	ENST00000256785	NM_030787.3	ENSP00000256785	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-32D17.4	ENST00000442280	?	1