Locus 195 (rs1789)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,565,736-15,695,363
block size	129,628 bp
variant count	45 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.773 [-5.238 – 1.138]	**gene(s) hit or close-by**	CC2D2A, FAM200B, FBXL5, RP11-799M12.2
phastCons	μ = 0.052 [0 – 0.697]	**eQTL gene(s)**	C1QTNF7, CC2D2A, FBXL5, RP11-484O2.1, RP11-799M12.2
GERP++	μ = -0.372 [-4.89 – 3.14]	**potentially regulated gene(s)**	BST1, FBXL5
CADD score	μ = 4.067 [0.078 – 12.67]	**disease gene(s)**	CC2D2A

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
palmitoyl sphingomyelin	6.85×10^{-5}	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
CC2D2A	COACH syndrome	DECIPHER	MIM:216360
CC2D2A	MECKEL SYNDROME, TYPE 6	DECIPHER	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	DECIPHER	MIM:612285
CC2D2A	MECKEL SYNDROME, TYPE 6	OMIM	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	OMIM	MIM:612285
CC2D2A	COACH SYNDROME	OMIM	MIM:216360
CC2D2A	Joubert syndrome with oculorenal defect	OrphaNet	OrphaNet:2318
CC2D2A	Meckel syndrome	OrphaNet	OrphaNet:564
CC2D2A	Joubert syndrome with hepatic defect	OrphaNet	OrphaNet:1454

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-799M12.2	?	ENSG00000273133	muscularis mucosae	2.95×10^{-6} (p-value)	GTEx Portal V6	10
RP11-799M12.2	?	ENSG00000273133	lung	2.20×10^{-7} (p-value)	GTEx Portal V6	42
RP11-799M12.2	?	ENSG00000273133	atrial appendage	6.02×10^{-8} (p-value)	GTEx Portal V6	43
RP11-799M12.2	?	ENSG00000273133	tibial nerve	1.21×10^{-6} (p-value)	GTEx Portal V6	35
CC2D2A	?	ENSG00000048342	blood	5.17×10^{-7} (p-value)	GTEx Portal V6	44
RP11-799M12.2	?	ENSG00000273133	blood	7.88×10^{-20} (p-value)	GTEx Portal V6	45
C1QTNF7	?	ENSG00000163145	blood	9.51×10^{-8} (p-value)	GTEx Portal V6	43
RP11-484O2.1	?	ENSG00000251379	blood	1.64×10^{-6} (p-value)	GTEx Portal V6	33
RP11-799M12.2	?	ENSG00000273133	tibial artery	1.05×10^{-9} (p-value)	GTEx Portal V6	45

FBXL5	?	ENSG00000118564	esophagus mucosa	2.34×10-7 (p-value)	GTEx Portal V6	43
FBXL5	?	ENSG00000118564	thyroid	2.81×10-9 (p-value)	GTEx Portal V6	45
RP11-799M12.2	?	ENSG00000273133	thyroid	2.07×10-7 (p-value)	GTEx Portal V6	45
RP11-799M12.2	?	ENSG00000273133	testis	6.83×10-9 (p-value)	GTEx Portal V6	44
RP11-799M12.2	?	ENSG00000273133	sun exposed skin	2.79×10-10 (p-value)	GTEx Portal V6	45
RP11-799M12.2	?	ENSG00000273133	subcutaneous adipocytes	1.67×10-15 (p-value)	GTEx Portal V6	45
FBXL5	ENST00000511441	ILMN_1673370	skin	5.90×10-11 (p-value)	MuTHER consortium	16
FBXL5	ENST00000412094		blood	1.34×10-36 (p-value)	MuTHER consortium	16
FBXL5	ENST00000513163		adipocyte	1.64×10-26 (p-value)	MuTHER consortium	16
FBXL5	ENST00000341285					
FBXL5	ENST00000511441	ILMN_1673370	monocyte	7.94×10-10 (p-value)	Fairfax et al.	6
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
FBXL5	ENST00000511441	209005_at	blood	1.00×10-12 (p-value)	Dixon et al.	4
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
FBXL5	ENST00000511441	ILMN_1673370	monocyte	7.68×10-103 (p-value)	Zeller et al.	2
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000374191	1	ENCP00000040472	FBXL5
		ENCP00000040476	BST1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001429713 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, H4K5ac, H3K36me3, H3K27me3, PolII, Rad21, TAF7, CTCF, H3K27ac
		HSMMtube	H3K4me3, H2AZ, DNase1, H3K79me2, CTCF, H3K9ac, H3K4me2, H3K27ac
		blood (K562)	H3K36me3, DNase1, H3K4me3, CTCF, H3K4me2, Egr1, H3K27ac, Max, Rad21, E2F6, H3K9ac, USF1, Cmyc, H3K79me2, H2AZ, PolII
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		liver (HepG2)	DNase1, PolII, H3K79me2, Rad21, TAF1, USF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3
		blood (GM12878)	CTCF, H3K9ac, PolII, H2AZ, DNase1, Egr1, Yy1, Rad21, H3K79me2, H3K4me3, USF1, H3K27ac, H3K4me2
		lung (IMR90)	DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K4me3, H3K4ac, H3K9ac, CTCF
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K27ac, H3K9ac
		NHLF	H3K4me3, DNase1, H3K9ac, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K36me3, H3K4me3, H3K9ac, H3K27ac
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, CTCF, PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3

		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
ENSR00001683631 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27ac, H3K4me2, H3K4me1
		A549	H3K36me3
		nervous (NH-A)	H3K36me3
ENSR00001429716 (enhancer)	2	monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		blood (DND-41)	H3K36me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3, H3K79me2
ENSR00001237226 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, H3K36me3, CTCF, PolII, Rad21, Gabp, TAF7, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, Srf
		HSMMtube	H3K4me3, H2AZ, DNase1, H3K4me2, H3K9ac, H3K79me2
		blood (K562)	Ini1, Egr1, H3K27ac, ZBTB33, Max, Srf, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K4me3, DNase1, H3K9ac
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2
		liver (HepG2)	H3K9ac, H3K27ac, H3K4me3, DNase1, HNF4A, H3K4me2, Gabp, Srf, Sin3Ak20, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, ZBTB33
		lung (IMR90)	H3K4me3, H3K9ac, DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H3K36me3
		blood (GM12878)	Sin3Ak20, PU1, PolII, H2AZ, DNase1, ELF1, H3K79me2, H3K4me3, ZBTB33, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K36me3, DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, Gabp, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K9ac, H3K36me3, H3K4me3, H3K27ac, DNase1, H3K4me1, H3K4me2
		endothelium (HUVEC)	PolII, DNase1, H3K36me3, H3K27ac, H3K9ac, Max, H3K4me2, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CC2D2A	downstream gene variant	3251	ENST00000503292	NM_001080522.2	ENSP00000421809	1
CC2D2A	downstream gene variant	3262	ENST00000389652	?	ENSP00000374303	1
CC2D2A	upstream gene variant, downstream gene variant	1711	ENST00000513035	?	?	3
CC2D2A	downstream gene variant	3437	ENST00000514039	?	?	1
CC2D2A	downstream gene variant	3251	ENST00000424120	?	ENSP00000403465	1
CC2D2A	downstream gene variant	3256	ENST00000506643	?	ENSP00000422931	1
FAM200B	upstream gene variant	938	ENST00000509022	?	?	2
FAM200B	upstream gene variant	855	ENST00000504823	?	?	2
FAM200B	upstream gene variant	942	ENST00000504598	?	?	2
FAM200B	upstream gene variant	1021	ENST00000503600	?	?	2
FAM200B	upstream gene variant	848	ENST00000510920	?	?	2
FAM200B	upstream gene variant	942	ENST00000512855	?	?	2
FAM200B	upstream gene variant	1011	ENST00000508567	?	?	2

FAM200B	upstream gene variant	982	ENST00000507992	?	?	2
FAM200B	upstream gene variant	992	ENST00000513053	?	?	2
FAM200B	upstream gene variant	977	ENST00000507305	?	?	2
FAM200B	upstream gene variant, downstream gene variant	904	ENST00000504137	?	?	3
FAM200B	upstream gene variant	1092	ENST00000510186	?	?	2
FAM200B	upstream gene variant	919	ENST00000515697	?	?	2
FAM200B	upstream gene variant	940	ENST00000510032	?	?	2
FAM200B	upstream gene variant, downstream gene variant	992	ENST00000422728	?	ENSP00000393017	3
FAM200B	upstream gene variant	943	ENST00000502502	?	?	2
FAM200B	upstream gene variant	1009	ENST00000514803	?	?	2
FAM200B	upstream gene variant, downstream gene variant	1047	ENST00000502856	?	?	3
FAM200B	upstream gene variant	942	ENST00000515430	?	?	2
FAM200B	upstream gene variant, downstream gene variant	915	ENST00000622362	NM_001145191.1	ENSP00000483930	3
FAM200B	upstream gene variant	943	ENST00000503617	?	ENSP00000422751	2
FAM200B	upstream gene variant, downstream gene variant	1593	ENST00000506610	?	?	4
FAM200B	upstream gene variant, downstream gene variant	942	ENST00000505260	?	?	3
FBXL5	downstream gene variant, upstream gene variant	198	ENST00000511441	?	ENSP00000425027	5
FBXL5	downstream gene variant, upstream gene variant	1410	ENST00000514541	?	?	4
FBXL5	downstream gene variant, upstream gene variant	1446	ENST00000507899	?	ENSP00000422049	4
FBXL5	downstream gene variant, upstream gene variant	1454	ENST00000515679	?	ENSP00000424423	4
FBXL5	downstream gene variant, upstream gene variant	3048	ENST00000509314	?	ENSP00000421070	4
FBXL5	downstream gene variant, upstream gene variant	1028	ENST00000512066	?	ENSP00000426993	4
FBXL5	downstream gene variant, upstream gene variant	335	ENST00000504837	?	?	5
FBXL5	downstream gene variant, upstream gene variant	3045	ENST00000503196	?	ENSP00000425541	4
FBXL5	downstream gene variant, upstream gene variant	1522	ENST00000510802	?	ENSP00000423961	3
FBXL5	downstream gene variant, upstream gene variant	289	ENST00000507700	?	?	5
FBXL5	downstream gene variant, upstream gene variant	198	ENST00000412094	NM_001193535.1	ENSP00000408679	5
FBXL5	downstream gene variant, upstream gene variant	198	ENST00000513163	?	ENSP00000425472	4
FBXL5	downstream gene variant, upstream gene variant	218	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CC2D2A	ENST00000506643	?	ENSP00000422931	7
CC2D2A	ENST00000503292	NM_001080522.2	ENSP00000421809	7
CC2D2A	ENST00000514039	?	?	3
CC2D2A	ENST00000389652	?	ENSP00000374303	7
CC2D2A	ENST00000424120	?	ENSP00000403465	7

CC2D2A	ENST00000424120	?	ENSP00000403465	7
FAM200B	ENST00000509022	?	?	1
FAM200B	ENST00000622362	NM_001145191.1	ENSP00000483930	1
FAM200B	ENST00000514803	?	?	1
FAM200B	ENST00000510920	?	?	1
FAM200B	ENST00000512855	?	?	1
FAM200B	ENST00000508567	?	?	1
FAM200B	ENST00000507992	?	?	1
FAM200B	ENST00000507305	?	?	1
FAM200B	ENST00000513053	?	?	1
FAM200B	ENST00000504137	?	?	1
FAM200B	ENST00000510186	?	?	1
FAM200B	ENST00000510032	?	?	1
FAM200B	ENST00000502856	?	?	1
FAM200B	ENST00000503600	?	?	1
FAM200B	ENST00000503617	?	ENSP00000422751	1
FAM200B	ENST00000515697	?	?	1
FAM200B	ENST00000502502	?	?	1
FAM200B	ENST00000422728	?	ENSP00000393017	1
FAM200B	ENST00000504598	?	?	2
FAM200B	ENST00000515430	?	?	1
FAM200B	ENST00000504823	?	?	1
FAM200B	ENST00000505260	?	?	1
FBXL5	ENST00000514541	?	?	9
FBXL5	ENST00000507899	?	ENSP00000422049	16
FBXL5	ENST00000515679	?	ENSP00000424423	16
FBXL5	ENST00000510802	?	ENSP00000423961	19
FBXL5	ENST00000509314	?	ENSP00000421070	10
FBXL5	ENST00000504837	?	?	7
FBXL5	ENST00000512066	?	ENSP00000426993	4
FBXL5	ENST00000503196	?	ENSP00000425541	10
FBXL5	ENST00000507700	?	?	21
FBXL5	ENST00000513163	?	ENSP00000425472	20
FBXL5	ENST00000412094	NM_001193535.1	ENSP00000408679	23
FBXL5	ENST00000511441	?	ENSP00000425027	23
FBXL5	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	23

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FBXL5	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

RP11-799M12.2	ENST00000609724	?	1