Locus 18 (rs2302465)

SNiPAcard

Block annotations

Block info

genomic range	chr4:15,527,099-15,730,349
block size	203,251 bp
variant count	78 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.038 [-4.028 – 5.711]	gene(s) hit or close-by	BST1, CC2D2A, FAM200B, FBXL5, RP11-115L11.1, RP11-799M12.2
phastCons	μ = 0.086 [0 – 0.995]	eQTL gene(s)	CD38, FAM200B, FBXL5, RP11-799M12.2
GERP++	μ = -0.416 [-7.17 – 5.53]	potentially regulated gene(s)	BST1, C1QTNF7, FAM200B, FBXL5, FGFBP2, RP11-442P12.1, RP11-442P12.2
CADD score	μ = 4.767 [0.048 – 24.4]	disease gene(s)	CC2D2A

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	conflicting interpretations of pathogenicity	ClinVar	RCV000079447.7	1
?	HGMD curated	HGMD	CM065042	1

Disease gene annotation

gene	trait	source DB	source entry/link
CC2D2A	MECKEL SYNDROME, TYPE 6	OMIM	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	OMIM	MIM:612285
CC2D2A	COACH SYNDROME	OMIM	MIM:216360
CC2D2A	COACH syndrome	DECIPHER	MIM:216360
CC2D2A	MECKEL SYNDROME, TYPE 6	DECIPHER	MIM:612284
CC2D2A	JOUBERT SYNDROME 9	DECIPHER	MIM:612285
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 transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
BST1	missense variant	ENST00000382346	?	ENSP00000371783	H/R	cAt/cGt	?	?	1
BST1	missense variant	ENST00000265016	NM_004334.2	ENSP00000265016	H/R	cAt/cGt	?	?	1
BST1	missense variant	ENST00000505785	?	ENSP00000423357	H/R	cAt/cGt	?	?	1
FAM200B	missense variant	ENST00000503617	?	ENSP00000422751	H/D	Cat/Gat	?	?	1
FAM200B	missense variant	ENST00000622362	NM_001145191.1	ENSP00000483930	H/D	Cat/Gat	?	?	1
FAM200B	missense variant	ENST00000422728	?	ENSP00000393017	H/D	Cat/Gat	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FBXL5	ENST00000511441	ILMN_1673370	blood	2.34×10^{-7} (p-value)	MuTHER consortium	7
FBXL5	ENST00000412094		adipocyte	4.43×10^{-5} (p-value)	MuTHER consortium	7
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
CD38	ENST00000502843	ILMN_2233783	adipocyte	1.11×10^{-8} (p-value)	MuTHER consortium	7
CD38	ENST00000226279					
FBXL5	ENST00000511441	ILMN_1673370	monocyte	3.60×10^{-19} (p-value)	Zeller et al.	2
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					
FAM200B	?	ENSG00000237765	transformed fibroblasts	6.06×10^{-6} (p-value)	GTEx Portal V6	43
FAM200B	?	ENSG00000237765	EBV lymphocytes	2.63×10^{-6} (p-value)	GTEx Portal V6	50
RP11-799M12.2	?	ENSG00000273133	blood	2.33×10^{-6} (p-value)	GTEx Portal V6	1
FBXL5	ENST00000511441	ILMN_1673370	monocyte	2.17×10^{-4} (p-value)	Fairfax et al.	1
FBXL5	ENST00000412094					
FBXL5	ENST00000513163					
FBXL5	ENST00000341285					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000040477		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000374034	1	ENCP00000040459	C1QTNF7
		ENCP00000040482	RP11-442P12.1
		ENCP00000040484	FGFBP2
		ENCP00000040479	RP11-442P12.2
ENCE00000374231	1	ENCP00000040475	BST1
ENCE00000374206	1	ENCP00000040473	FBXL5 FAM200B
ENCE00000374192	1	ENCP00000040472	FBXL5
		ENCP00000040476	BST1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001237209 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	H3K36me3
ENSR00001237214 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3
	5	embryonic stem cell (H1ESC)	DNase1, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, H4K5ac, H3K36me3, H3K27me3,

ENSR00001429713 (promoter)			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
ENSR00001429714 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3
		A549	H3K36me3
ENSR00001429715 (promoter flanking region)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	DNase1, H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	DNase1, H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K27ac, Jund
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	Cjun
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		nervous (NH-A)	DNase1, H3K36me3
		skin (NHEK)	DNase1, H3K4me1, H3K36me3
ENSR00001237226 (promoter)	2	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
ENSR00001237231	1	NHLF	H3K27ac, DNase1

(promoter flanking region)		embryonic stem cell (H1ESC) HSMMtube	DNase1, H3K36me3 H3K27ac, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K36me3, H3K4me1, DNase1
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3, H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		lung (IMR90)	DNase1, H3K18ac, H3K36me3, H2BK120ac
		nervous (NH-A)	DNase1, H3K9ac, H3K27ac, H3K36me3
		A549	H3K4me3, DNase1, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me2, H3K27ac, H3K4me1, DNase1
ENSR00001237239 (enhancer)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00001429723 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac, H3K36me3
		HSMMtube	DNase1
		nervous (NH-A)	H3K36me3, DNase1
		blood (K562)	H3K27me3
		A549	H3K36me3
		muscle (HSMM)	DNase1, H3K4me2, H3K27ac, H2AZ

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BST1	upstream gene variant	2095	ENST00000514989	?	ENSP00000424761	3
BST1	upstream gene variant	2212	ENST00000505785	?	ENSP00000423357	1
BST1	upstream gene variant	1461	ENST00000382346	?	ENSP00000371783	2
BST1	upstream gene variant	575	ENST00000514445	?	ENSP00000420925	2
BST1	upstream gene variant	1315	ENST00000265016	NM_004334.2	ENSP00000265016	2
CC2D2A	downstream gene variant	1001	ENST00000512202	?	?	5
CC2D2A	downstream gene variant	1080	ENST00000512702	?	ENSP00000422875	1
CC2D2A	downstream gene variant	991	ENST00000389652	?	ENSP00000374303	2
CC2D2A	upstream gene variant, downstream gene variant	1275	ENST00000513035	?	?	2
CC2D2A	downstream gene variant	1166	ENST00000514039	?	?	2
CC2D2A	downstream gene variant	985	ENST00000506643	?	ENSP00000422931	2
CC2D2A	upstream gene variant, downstream gene variant	2143	ENST00000510220	?	?	2
CC2D2A	downstream gene variant	980	ENST00000424120	?	ENSP00000403465	2
CC2D2A	downstream gene variant	980	ENST00000503292	NM_001080522.2	ENSP00000421809	2
FAM200B	upstream gene variant, downstream gene variant	375	ENST00000509022	?	?	6
FAM200B	upstream gene variant, downstream gene variant	964	ENST00000422728	?	ENSP00000393017	5
FAM200B	upstream gene variant, downstream gene variant	915	ENST00000503617	?	ENSP00000422751	5
FAM200B	upstream gene variant, downstream gene variant	914	ENST00000505260	?	?	5
FAM200B	upstream gene variant, downstream gene variant	914	ENST00000515430	?	?	5
FAM200B	upstream gene variant, downstream gene variant	529	ENST00000506610	?	?	5
FAM200B	upstream gene variant, downstream gene variant	1064	ENST00000510186	?	?	4

FAM200B	upstream gene variant, downstream gene variant	1019	ENST00000502856	?	?	4
FAM200B	upstream gene variant, downstream gene variant	413	ENST00000502502	?	?	6
FAM200B	upstream gene variant, downstream gene variant	891	ENST00000515697	?	?	5
FAM200B	upstream gene variant, downstream gene variant	993	ENST00000503600	?	?	5
FAM200B	upstream gene variant, downstream gene variant	983	ENST00000508567	?	?	5
FAM200B	upstream gene variant, downstream gene variant	827	ENST00000504823	?	?	5
FAM200B	upstream gene variant, downstream gene variant	375	ENST00000514803	?	?	6
FAM200B	upstream gene variant, downstream gene variant	914	ENST00000504598	?	?	5
FAM200B	upstream gene variant, downstream gene variant	646	ENST00000622362	NM_001145191.1	ENSP00000483930	5
FAM200B	upstream gene variant, downstream gene variant	914	ENST00000512855	?	?	5
FAM200B	upstream gene variant, downstream gene variant	820	ENST00000510920	?	?	5
FAM200B	upstream gene variant, downstream gene variant	638	ENST00000504137	?	?	5
FAM200B	upstream gene variant, downstream gene variant	585	ENST00000507992	?	?	6
FAM200B	upstream gene variant, downstream gene variant	949	ENST00000507305	?	?	5
FAM200B	upstream gene variant, downstream gene variant	375	ENST00000510032	?	?	6
FAM200B	upstream gene variant, downstream gene variant	964	ENST00000513053	?	?	5
FBXL5	downstream gene variant, upstream gene variant	414	ENST00000512066	?	ENSP00000426993	5
FBXL5	downstream gene variant, upstream gene variant	154	ENST00000507700	?	?	3
FBXL5	downstream gene variant, upstream gene variant	999	ENST00000515679	?	ENSP00000424423	6
FBXL5	downstream gene variant, upstream gene variant	2083	ENST00000514541	?	?	5
FBXL5	downstream gene variant, upstream gene variant	991	ENST00000507899	?	ENSP00000422049	6
FBXL5	downstream gene variant, upstream gene variant	349	ENST00000509314	?	ENSP00000421070	3
FBXL5	downstream gene variant, upstream gene variant	339	ENST00000503196	?	ENSP00000425541	3
FBXL5	downstream gene variant, upstream gene variant	606	ENST00000412094	NM_001193535.1	ENSP00000408679	4
FBXL5	downstream gene variant, upstream gene variant	490	ENST00000510802	?	ENSP00000423961	4
FBXL5	downstream gene variant, upstream gene variant	139	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	4
FBXL5	downstream gene variant, upstream gene variant	443	ENST00000513163	?	ENSP00000425472	6
FBXL5	downstream gene variant, upstream gene variant	606	ENST00000511441	?	ENSP00000425027	4
FBXL5	downstream gene variant, upstream gene variant	718	ENST00000504837	?	?	3
RP11-115L11.1	downstream gene variant	2236	ENST00000498116	?	?	1
RP11-799M12.2	downstream gene variant	2495	ENST00000609724	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BST1	ENST00000265016	NM_004334.2	ENSP00000265016	7
BST1	ENST00000505785	?	ENSP00000423357	5

BST1	ENST00000382346	?	ENSP00000371783	7
BST1	ENST00000514989	?	ENSP00000424761	3
BST1	ENST00000514445	?	ENSP00000420925	5
CC2D2A	ENST00000512202	?	?	1
CC2D2A	ENST00000512702	?	ENSP00000422875	2
CC2D2A	ENST00000389652	?	ENSP00000374303	14
CC2D2A	ENST00000514039	?	?	2
CC2D2A	ENST00000424120	?	ENSP00000403465	14
CC2D2A	ENST00000513811	?	?	3
CC2D2A	ENST00000510220	?	?	1
CC2D2A	ENST00000503292	NM_001080522.2	ENSP00000421809	14
CC2D2A	ENST00000506643	?	ENSP00000422931	12
FAM200B	ENST00000509022	?	?	3
FAM200B	ENST00000422728	?	ENSP00000393017	3
FAM200B	ENST00000505260	?	?	4
FAM200B	ENST00000503617	?	ENSP00000422751	3
FAM200B	ENST00000506610	?	?	1
FAM200B	ENST00000503600	?	?	4
FAM200B	ENST00000510920	?	?	4
FAM200B	ENST00000502502	?	?	3
FAM200B	ENST00000515697	?	?	4
FAM200B	ENST00000512855	?	?	4
FAM200B	ENST00000507992	?	?	3
FAM200B	ENST00000514803	?	?	3
FAM200B	ENST00000622362	NM_001145191.1	ENSP00000483930	3
FAM200B	ENST00000515430	?	?	4
FAM200B	ENST00000504823	?	?	4
FAM200B	ENST00000504598	?	?	8
FAM200B	ENST00000507305	?	?	4
FAM200B	ENST00000508567	?	?	4
FAM200B	ENST00000502856	?	?	4
FAM200B	ENST00000510032	?	?	3
FAM200B	ENST00000510186	?	?	4
FAM200B	ENST00000504137	?	?	4
FAM200B	ENST00000513053	?	?	4
FBXL5	ENST00000509314	?	ENSP00000421070	17
FBXL5	ENST00000504837	?	?	16
FBXL5	ENST00000514541	?	?	13
FBXL5	ENST00000507899	?	ENSP00000422049	27
FBXL5	ENST00000515679	?	ENSP00000424423	27
FBXL5	ENST00000510802	?	ENSP00000423961	29
FBXL5	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	33

FBXL5	ENST00000412094	NM_001193535.1	ENSP00000408679	33
FBXL5	ENST00000507700	?	?	32
FBXL5	ENST00000503196	?	ENSP00000425541	17
FBXL5	ENST00000513163	?	ENSP00000425472	23
FBXL5	ENST00000512066	?	ENSP00000426993	6
FBXL5	ENST00000511441	?	ENSP00000425027	33

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FBXL5	ENST00000509314	?	ENSP00000421070	1
FBXL5	ENST00000507899	?	ENSP00000422049	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FAM200B	ENST00000505260	?	1
FAM200B	ENST00000512855	?	1
FAM200B	ENST00000515430	?	1
FAM200B	ENST00000504137	?	1
FBXL5	ENST00000514541	?	1