SNiPAcard

Block annotations

Block info

genomic range	chr4:15,551,298-15,719,439
block size	168,142 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.028 [-3.15 – 3.645]	**gene(s) hit or close-by**	BST1, CC2D2A, FAM200B, FBXL5
phastCons	μ = 0.184 [0 – 1]	**eQTL gene(s)**	BST1
GERP++	μ = 0.117 [-2.61 – 3.84]	**potentially regulated gene(s)**	BST1, FBXL5
CADD score	μ = 5.313 [0.018 – 18.87]	**disease gene(s)**	CC2D2A

Trait annotations

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	Q/R	cAa/cGa	?	?	1
BST1	missense variant	ENST00000265016	NM_004334.2	ENSP00000265016	Q/R	cAa/cGa	?	?	1
BST1	missense variant	ENST00000505785	?	ENSP00000423357	Q/R	cAa/cGa	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
BST1	ENST00000382346	ILMN_1770161	monocyte	1.56×10-4 (p-value)	Fairfax et al.	2
BST1	ENST00000265016					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000634801	1	ENST00000510802, ENST00000515679, ENST00000503196, ENST00000509314	FBXL5

ENCODE promoter-associated DHS

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

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001237239 (enhancer)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00001237240 (promoter flanking region)	2	NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		Osteobl	H3K27ac
		nervous (NH-A)	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	4309	ENST00000265016	NM_004334.2	ENSP00000265016	1
BST1	upstream gene variant	846	ENST00000514989	?	ENSP00000424761	3
BST1	upstream gene variant	4177	ENST00000514445	?	ENSP00000420925	1
BST1	upstream gene variant	4455	ENST00000382346	?	ENSP00000371783	1
CC2D2A	upstream gene variant	1063	ENST00000512202	?	?	1
FAM200B	upstream gene variant	954	ENST00000506610	?	?	1
FAM200B	downstream gene variant	2064	ENST00000504598	?	?	1
FBXL5	upstream gene variant	1517	ENST00000507700	?	?	4
FBXL5	upstream gene variant	2174	ENST00000507899	?	ENSP00000422049	1
FBXL5	upstream gene variant	640	ENST00000512066	?	ENSP00000426993	1
FBXL5	downstream gene variant, upstream gene variant	526	ENST00000514541	?	?	5
FBXL5	upstream gene variant	1476	ENST00000412094	NM_001193535.1	ENSP00000408679	4
FBXL5	upstream gene variant	2161	ENST00000510802	?	ENSP00000423961	1
FBXL5	upstream gene variant	1517	ENST00000511441	?	ENSP00000425027	4
FBXL5	upstream gene variant	1563	ENST00000504837	?	?	4
FBXL5	upstream gene variant	669	ENST00000513163	?	ENSP00000425472	1
FBXL5	upstream gene variant	2109	ENST00000515679	?	ENSP00000424423	1
FBXL5	upstream gene variant	1446	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
BST1	ENST00000382346	?	ENSP00000371783	S	tcT/tcC	1
BST1	ENST00000265016	NM_004334.2	ENSP00000265016	S	tcT/tcC	1
BST1	ENST00000514445	?	ENSP00000420925	S	tcT/tcC	1
BST1	ENST00000505785	?	ENSP00000423357	S	tcT/tcC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BST1	ENST00000382346	?	ENSP00000371783	3
BST1	ENST00000265016	NM_004334.2	ENSP00000265016	3
BST1	ENST00000514445	?	ENSP00000420925	3
BST1	ENST00000514989	?	ENSP00000424761	1
BST1	ENST00000505785	?	ENSP00000423357	3
CC2D2A	ENST00000503292	NM_001080522.2	ENSP00000421809	2
CC2D2A	ENST00000389652	?	ENSP00000374303	2
CC2D2A	ENST00000424120	?	ENSP00000403465	2
CC2D2A	ENST00000506643	?	ENSP00000422931	2
CC2D2A	ENST00000512202	?	?	1
FAM200B	ENST00000622362	NM_001145191.1	ENSP00000483930	1
FAM200B	ENST00000422728	?	ENSP00000393017	1
FAM200B	ENST00000503617	?	ENSP00000422751	1
FAM200B	ENST00000505260	?	?	1
FAM200B	ENST00000503600	?	?	1
FAM200B	ENST00000510920	?	?	1
FAM200B	ENST00000509022	?	?	1
FAM200B	ENST00000514803	?	?	1
FAM200B	ENST00000515697	?	?	1
FAM200B	ENST00000502502	?	?	1
FAM200B	ENST00000512855	?	?	1
FAM200B	ENST00000504598	?	?	2
FAM200B	ENST00000504823	?	?	1
FAM200B	ENST00000515430	?	?	1
FAM200B	ENST00000502856	?	?	1
FAM200B	ENST00000510186	?	?	1
FAM200B	ENST00000510032	?	?	1
FAM200B	ENST00000508567	?	?	1
FAM200B	ENST00000507305	?	?	1
FAM200B	ENST00000513053	?	?	1
FAM200B	ENST00000507992	?	?	1
FAM200B	ENST00000504137	?	?	1

FBXL5	ENST00000515679	?	ENSP00000424423	7
FBXL5	ENST00000509314	?	ENSP00000421070	5
FBXL5	ENST00000507899	?	ENSP00000422049	7
FBXL5	ENST00000514541	?	?	2
FBXL5	ENST00000510802	?	ENSP00000423961	7
FBXL5	ENST00000507700	?	?	3
FBXL5	ENST00000412094	NM_001193535.1	ENSP00000408679	3
FBXL5	ENST00000341285	NM_012161.3, NM_001193534.1	ENSP00000344866	3
FBXL5	ENST00000504837	?	?	1
FBXL5	ENST00000513163	?	ENSP00000425472	2
FBXL5	ENST00000503196	?	ENSP00000425541	5
FBXL5	ENST00000511441	?	ENSP00000425027	3