Locus 38 (rs3742264)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,602,162-46,655,449
block size	53,288 bp
variant count	63 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.509 [-2.944 – 1.624]	gene(s) hit or close-by	CPB2, CPB2-AS1, ZC3H13
phastCons	μ = 0.064 [0 – 1]	eQTL gene(s)	CPB2, CPB2-AS1, ZC3H13
GERP++	μ = -0.460 [-5.92 – 2.82]	potentially regulated gene(s)	LCP1, LRRC63
CADD score	μ = 4.005 [0.041 – 18.87]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR080756	1
?	HGMD curated	HGMD	CM063919	1

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)
CPB2	missense variant	ENST00000439329	NM_001278541.1	ENSP00000400714	T/A	Aca/Gca	?	?	1
CPB2	missense variant	ENST00000181383	NM_001872.4	ENSP00000181383	T/A	Aca/Gca	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CPB2-AS1	?	ENSG00000235903	nucleus accumbens	5.04×10^{-10} (p-value)	GTEx Portal V6	62
ZC3H13	?	ENSG00000123200	sun exposed skin	5.31×10^{-7} (p-value)	GTEx Portal V6	43
CPB2	?	ENSG00000080618	sun exposed skin	7.22×10^{-6} (p-value)	GTEx Portal V6	1
CPB2-AS1	?	ENSG00000235903	thyroid	4.23×10^{-8} (p-value)	GTEx Portal V6	61
CPB2-AS1	?	ENSG00000235903	cerebellum	2.92×10^{-8} (p-value)	GTEx Portal V6	59
CPB2-AS1	?	ENSG00000235903	cerebellar hemisphere	4.23×10^{-6} (p-value)	GTEx Portal V6	53
CPB2-AS1	?	ENSG00000235903	pituitary	3.78×10^{-7} (p-value)	GTEx Portal V6	54
CPB2-AS1	?	ENSG00000235903	caudate basal ganglia	1.48×10^{-6} (p-value)	GTEx Portal V6	4
CPB2-AS1	?	ENSG00000235903	liver	2.97×10^{-6} (p-value)	GTEx Portal V6	1

CPB2-AS1	?	ENSG00000235903	lung	4.50×10-7 (p-value)	GTEx Portal V6	4
CPB2-AS1	?	ENSG00000235903	hypothalamus	3.14×10-7 (p-value)	GTEx Portal V6	2
CPB2-AS1	?	ENSG00000235903	sigmoid colon	2.59×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000157095	2	ENCP00000018502	LCP1
		ENCP00000018503	LRRC63 LCP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000515658 (promoter)	1	embryonic stem cell (H1ESC)	CTCF, PolII, Gabp, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K9ac, H3K79me2, H3K27ac, DNase1, H2AZ, H3K4me3
		blood (K562)	H3K4me3, H3K4me2, PolII, TAF1, H2AZ, H3K79me2, Gabp, H3K9ac, CTCF, Max, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K9ac, DNase1, H3K4me2, H3K4me3, H2AZ, H3K79me2
		liver (HepG2)	H3K27me3, DNase1, H3K36me3, Gabp, PolII, H3K79me2, ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3
		lung (IMR90)	H3K4me2, H3K4me3, H3K9ac, DNase1
		blood (GM12878)	DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H2AZ, PolII, CTCF, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K9ac, H3K4me3, DNase1, H3K27ac
		Osteobl	H3K4me3, H3K4me2, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac, DNase1, PolII, Gabp, H3K79me2
		monocytes (Monocytes-CD14+)	H3K9ac, H3K27me3, H3K4me3, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	Max, H3K4me2, H3K4me3, H3K9ac, PolII, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001509498 (CTCF binding site)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	Rad21, CTCF, USF1
		liver (HepG2)	H3K36me3, Rad21
		HSMMtube	H3K27me3
		blood (GM12878)	BATF, MEF2A, H2AZ, Rad21, H3K27ac, CTCF
		nervous (NH-A)	DNase1
		blood (K562)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ZC3H13	upstream gene variant	558	ENST00000242848	?	ENSP00000242848	1
ZC3H13	upstream gene variant	619	ENST00000428921	?	ENSP00000416238	1
ZC3H13	upstream gene variant	545	ENST00000282007	NM_015070.4	ENSP00000282007	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	D	gaC/gaT	1

Intron variant

intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	58
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	58
CPB2-AS1	ENST00000606991	?	?	59
CPB2-AS1	ENST00000606351	?	?	59
CPB2-AS1	ENST00000415033	?	?	59
CPB2-AS1	ENST00000606243	?	?	59
ZC3H13	ENST00000428921	?	ENSP00000416238	2
ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	4
ZC3H13	ENST00000242848	?	ENSP00000242848	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	1
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	1