Locus 117 (rs9534288)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,569,846-46,636,652
block size	66,807 bp
variant count	70 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.506 [-3.814 – 2.895]	**gene(s) hit or close-by**	CPB2, CPB2-AS1, ZC3H13
phastCons	μ = 0.033 [0 – 0.978]	**eQTL gene(s)**	CPB2-AS1, LCP1
GERP++	μ = -0.155 [-5.96 – 3.84]	**potentially regulated gene(s)**	LCP1, LRRC63
CADD score	μ = 3.899 [0.032 – 14.11]	**disease gene(s)**	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR080759	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LCP1	ENST00000323076	ILMN_1662932	blood	6.46×10-6 (p-value)	Westra et al.	23
LCP1	ENST00000398576					
CPB2-AS1	?	ENSG00000235903	EBV lymphocytes	4.98×10-6 (p-value)	GTEx Portal V6	13

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000156965	1	ENCP00000018501	LCP1
ENCE00000156967	1	ENCP00000018501	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
ENSR00000515659 (promoter flanking region)	3	liver (HepG2)	H3K36me3
		skin (NHDF-AD)	DNase1
		skin (NHEK)	DNase1
		breast (HMEC)	H3K27ac
ENSR00001509497 (promoter flanking region)	2	liver (HepG2)	H3K36me3
		Osteobl	H3K27ac, H3K4me2
		A549	H3K4me3
		skin (NHEK)	H3K4me2
ENSR00001618259 (CTCF binding site)	1	liver (HepG2)	H3K36me3
		Osteobl	H3K27ac, H3K4me2
		A549	H3K4me3
		skin (NHEK)	H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CPB2	downstream gene variant	1735	ENST00000181383	NM_001872.4	ENSP00000181383	5
CPB2	downstream gene variant	1736	ENST00000439329	NM_001278541.1	ENSP00000400714	5
CPB2-AS1	upstream gene variant	1397	ENST00000606243	?	?	6
CPB2-AS1	upstream gene variant	1451	ENST00000415033	?	?	6
CPB2-AS1	upstream gene variant	1397	ENST00000606991	?	?	6
CPB2-AS1	upstream gene variant	1355	ENST00000606351	?	?	6
ZC3H13	upstream gene variant	599	ENST00000242848	?	ENSP00000242848	4
ZC3H13	downstream gene variant, upstream gene variant	981	ENST00000470308	?	?	11
ZC3H13	downstream gene variant, upstream gene variant	594	ENST00000464597	?	ENSP00000480658	10
ZC3H13	downstream gene variant, upstream gene variant	660	ENST00000428921	?	ENSP00000416238	7
ZC3H13	upstream gene variant	586	ENST00000282007	NM_015070.4	ENSP00000282007	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	15
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	15
CPB2-AS1	ENST00000606351	?	?	15
CPB2-AS1	ENST00000606243	?	?	15
CPB2-AS1	ENST00000606991	?	?	15
CPB2-AS1	ENST00000415033	?	?	15
ZC3H13	ENST00000464597	?	ENSP00000480658	7
ZC3H13	ENST00000470308	?	?	10
ZC3H13	ENST00000428921	?	ENSP00000416238	14
ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	55

ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	55
ZC3H13	ENST00000242848	?	ENSP00000242848	55

3'-UTR variant

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