Locus 8 (rs1926447)

SNiPAcard

Block annotations

Block info

genomic range	chr13:46,514,506-46,629,995
block size	115,490 bp
variant count	72 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.652 [-3.814 – 2.601]	gene(s) hit or close-by	CPB2, CPB2-AS1, ZC3H13
phastCons	μ = 0.045 [0 – 0.997]	eQTL gene(s)	LCP1
GERP++	μ = -0.574 [-7.55 – 3]	potentially regulated gene(s)	–
CADD score	μ = 3.565 [0.018 – 11.48]	disease gene(s)	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM013697	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	I/T	aTt/aCt	tolerated	benign	1
CPB2	missense variant	ENST00000181383	NM_001872.4	ENSP00000181383	I/T	aTt/aCt	tolerated	benign	1
ZC3H13	missense variant	ENST00000242848	?	ENSP00000242848	E/D	gaA/gaT	?	unknown	1
ZC3H13	missense variant	ENST00000282007	NM_015070.4	ENSP00000282007	E/D	gaA/gaT	?	unknown	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LCP1	ENST00000323076	ILMN_1662932	blood	5.07×10^{-6} (p-value)	Westra et al.	31
LCP1	ENST00000398576					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001509491 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3

	monocytes (Monocytes-CD14+)	H3K36me3
	liver (HepG2)	H3K36me3
	lung (IMR90)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ZC3H13	downstream gene variant, upstream gene variant	594	ENST00000464597	?	ENSP00000480658	13
ZC3H13	downstream gene variant, upstream gene variant	961	ENST00000282007	NM_015070.4	ENSP00000282007	7
ZC3H13	downstream gene variant, upstream gene variant	721	ENST00000242848	?	ENSP00000242848	4
ZC3H13	downstream gene variant, upstream gene variant	981	ENST00000470308	?	?	11
ZC3H13	downstream gene variant, upstream gene variant	2405	ENST00000428921	?	ENSP00000416238	7

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	H	caT/caC	1
ZC3H13	ENST00000242848	?	ENSP00000242848	H	caT/caC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPB2	ENST00000439329	NM_001278541.1	ENSP00000400714	2
CPB2	ENST00000181383	NM_001872.4	ENSP00000181383	2
CPB2-AS1	ENST00000606991	?	?	3
CPB2-AS1	ENST00000606243	?	?	3
CPB2-AS1	ENST00000606351	?	?	3
CPB2-AS1	ENST00000415033	?	?	3
ZC3H13	ENST00000464597	?	ENSP00000480658	9
ZC3H13	ENST00000470308	?	?	13
ZC3H13	ENST00000428921	?	ENSP00000416238	3
ZC3H13	ENST00000282007	NM_015070.4	ENSP00000282007	59
ZC3H13	ENST00000242848	?	ENSP00000242848	63

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ZC3H13	ENST00000470308	?	1