Locus 396 (rs3743392)

SNiPAcard

Block annotations

Block info

genomic range	chr15:89,436,085-89,449,137
block size	13,053 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.677 [-2.626 – 1.105]	**gene(s) hit or close-by**	HAPLN3, MFGE8
phastCons	μ = 0.015 [0 – 0.2]	**eQTL gene(s)**	HAPLN3, MFGE8
GERP++	μ = -0.415 [-2.92 – 1.63]	**potentially regulated gene(s)**	MFGE8
CADD score	μ = 3.569 [0.602 – 8.556]	**disease gene(s)**	–

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)
HAPLN3	stop lost	ENST00000562889	?	ENSP00000457180	*/Q	Tag/Cag	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HAPLN3	ENST00000558770	ILMN_1654319	blood	2.57×10^{-6} (p-value)	Westra et al.	6
HAPLN3	ENST00000359595					
MFGE8	?	ENSG00000140545	lung	9.36×10^{-10} (p-value)	GTEx Portal V6	12
MFGE8	?	ENSG00000140545	tibial nerve	7.12×10^{-8} (p-value)	GTEx Portal V6	4
MFGE8	?	ENSG00000140545	transformed fibroblasts	6.07×10^{-9} (p-value)	GTEx Portal V6	12
MFGE8	?	ENSG00000140545	esophagus mucosa	5.76×10^{-6} (p-value)	GTEx Portal V6	3
MFGE8	?	ENSG00000140545	left ventricle	3.44×10^{-6} (p-value)	GTEx Portal V6	5
MFGE8	?	ENSG00000140545	testis	2.47×10^{-6} (p-value)	GTEx Portal V6	4

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000215481	1	ENST00000268151, ENST00000558018, ENST00000268150	MFGE8
FFCP00000215480	1	ENST00000558018, ENST00000268151, ENST00000268150	MFGE8

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000413115	1	embryonic stem cell (H1ESC)	PolII, H3K36me3, DNase1

(TF binding site)	lung (IMR90)	H3K36me3
	A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HAPLN3	upstream gene variant	3688	ENST00000563808	?	?	2
HAPLN3	upstream gene variant	3668	ENST00000562281	?	ENSP00000456985	2
HAPLN3	upstream gene variant	3575	ENST00000359595	NM_178232.2	ENSP00000352606	2
HAPLN3	upstream gene variant	3670	ENST00000558770	?	ENSP00000456458	2
HAPLN3	upstream gene variant	3604	ENST00000562889	?	ENSP00000457180	2
MFGE8	downstream gene variant	114	ENST00000542878	?	ENSP00000444332	1
MFGE8	downstream gene variant	2113	ENST00000613965	?	ENSP00000478952	2
MFGE8	downstream gene variant	157	ENST00000558352	?	?	10
MFGE8	downstream gene variant, upstream gene variant	97	ENST00000560937	?	?	11
MFGE8	downstream gene variant, upstream gene variant	1260	ENST00000617199	?	?	3
MFGE8	downstream gene variant	4298	ENST00000558773	?	?	1
MFGE8	downstream gene variant	709	ENST00000559770	?	?	10
MFGE8	downstream gene variant	1521	ENST00000559259	?	?	9
MFGE8	downstream gene variant	1538	ENST00000559997	?	?	9
MFGE8	downstream gene variant, upstream gene variant	4	ENST00000560553	?	?	11
MFGE8	downstream gene variant	4874	ENST00000557944	?	?	1
MFGE8	downstream gene variant	1481	ENST00000558029	?	ENSP00000452926	2
MFGE8	downstream gene variant	3886	ENST00000559143	?	?	1

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
MFGE8	ENST00000558029	?	ENSP00000452926	1
MFGE8	ENST00000268151	NM_001114614.1	ENSP00000268151	1
MFGE8	ENST00000542878	?	ENSP00000444332	1
MFGE8	ENST00000558018	?	ENSP00000452734	1
MFGE8	ENST00000613965	?	ENSP00000478952	1
MFGE8	ENST00000268150	NM_005928.2	ENSP00000268150	1
MFGE8	ENST00000566497	?	ENSP00000456281	1
MFGE8	ENST00000559259	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HAPLN3	ENST00000359595	NM_178232.2	ENSP00000352606	2
HAPLN3	ENST00000558770	?	ENSP00000456458	2
HAPLN3	ENST00000563808	?	?	2
HAPLN3	ENST00000562889	?	ENSP00000457180	1
HAPLN3	ENST00000562281	?	ENSP00000456985	2

MFGE8	ENST00000560937	?	?	1
MFGE8	ENST00000559997	?	?	1
MFGE8	ENST00000558029	?	ENSP00000452926	10
MFGE8	ENST00000268151	NM_001114614.1	ENSP00000268151	12
MFGE8	ENST00000542878	?	ENSP00000444332	11
MFGE8	ENST00000558018	?	ENSP00000452734	12
MFGE8	ENST00000613965	?	ENSP00000478952	10
MFGE8	ENST00000268150	NM_005928.2	ENSP00000268150	12
MFGE8	ENST00000566497	?	ENSP00000456281	12

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MFGE8	ENST00000268151	NM_001114614.1	ENSP00000268151	1
MFGE8	ENST00000558018	?	ENSP00000452734	1
MFGE8	ENST00000268150	NM_005928.2	ENSP00000268150	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
MFGE8	ENST00000617199	?	9
MFGE8	ENST00000560553	?	1