Locus 192 (rs4494114)

SNiPAcard

Block annotations

Block info

genomic range	chr1:39,297,734-39,380,307
block size	82,574 bp
variant count	59 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.217 [-3.141 – 2.757]	gene(s) hit or close-by	GJA9, MYCBP, RHBDL2, RP5-864K19.4, RP5-864K19.6, RP5-864K19.7, RRAGC
phastCons	μ = 0.074 [0 – 1]	eQTL gene(s)	GJA9, MYCBP, RHBDL2, RP5-864K19.6, RP5-864K19.7
GERP++	μ = -0.291 [-6.75 – 6.03]	potentially regulated gene(s)	MACF1, RRAGC
CADD score	μ = 4.507 [0.029 – 20.2]	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)
GJA9	missense variant	ENST00000357771	NM_030772.4	ENSP00000350415	I/V	Ata/Gta	?	?	1
GJA9	missense variant	ENST00000360786	?	ENSP00000354020	I/V	Ata/Gta	?	?	1
RHBDL2	missense variant	ENST00000289248	?	ENSP00000289248	M/L	Atg/Ctg	?	?	1
RHBDL2	missense variant	ENST00000372990	NM_017821.3, NM_001304746.1	ENSP00000362081	M/L	Atg/Ctg	?	?	1
RP5-864K19.6	missense variant	ENST00000454994	?	ENSP00000406846	Q/R	cAg/cGg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RHBDL2	?	ENSG00000158315	esophagus mucosa	2.02×10^{-6} (p-value)	GTEx Portal V6	36
RP5-864K19.6	ENST00000489803	ILMN_1710161	blood	1.14×10^{-4} (p-value)	Westra et al.	1
GJA9	ENST00000357771					
RP5-864K19.7	ENST00000622355					
MYCBP	ENST00000397572					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
HNF4A	MA0114.2	6	yes	0.000	1
EGR1	MA0162.2	13	no	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000015074	1	ENCP00000001927	MACF1
		ENCP00000001918	RRAGC

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001579011 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, Jund, Sin3Ak20, H3K9ac, ATF3, TAF1, H3K4me2, SP1, USF1, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	Egr1, H3K27ac, Max, SP2, H3K9ac, Nfe2, USF1, Cmyc, H2AZ, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	TAF1, USF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, PolII, ATF3, DNase1
		blood (GM12878)	DNase1, Egr1, H3K79me2, H3K4me3, USF1, Cfos, H3K4me2, H3K9ac, Pbx3, ATF3, PolII, H2AZ
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K4me1, H3K4me2, H3K27ac
		breast (HMEC)	H3K4me3, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, Max, H3K4me3, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, PolII, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00000164746 (promoter)	2	embryonic stem cell (H1ESC)	H3K9ac, H3K36me3, CTCF, PolII, Rad21, Sin3Ak20, Yy1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K36me3, DNase1, H2AZ, H3K4me3, H3K4me2
		blood (K562)	H3K4me3, DNase1, H3K27ac, Max, H3K9ac, Gabp, H2AZ, PolII, H3K4me2, H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H2AZ, H3K4me3, H3K4me2
		liver (HepG2)	Gabp, PolII, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K4me2, H3K9ac, H3K4me3, DNase1
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	Nrf1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, Gabp, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me3, H3K4me2, PolII, DNase1
		A549	H3K36me3, DNase1, H3K4me3, H3K4me2, H3K9ac
ENSR00000281600 (CTCF binding site)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		liver (HepG2)	FOXA1, CTCF
		nervous (NH-A)	DNase1
		blood (K562)	Egr1, CTCF
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GJA9	downstream gene variant, upstream gene variant	541	ENST00000357771	NM_030772.4	ENSP00000350415	5
GJA9	downstream gene variant, upstream gene variant	441	ENST00000360786	?	ENSP00000354020	10
MYCBP	downstream gene variant, upstream gene variant	499	ENST00000495043	?	?	7
MYCBP	downstream gene variant, upstream gene variant	505	ENST00000397572	NM_012333.4	ENSP00000380702	7
MYCBP	downstream gene variant, upstream gene variant	409	ENST00000494695	?	ENSP00000485717	7
MYCBP	downstream gene variant, upstream gene variant	1172	ENST00000465771	?	?	7

RHBDL2	downstream gene variant	868	ENST00000540558	?	ENSP00000441097	4
RHBDL2	downstream gene variant	888	ENST00000372990	NM_017821.3, NM_001304746.1	ENSP00000362081	3
RHBDL2	downstream gene variant	1377	ENST00000289248	?	ENSP00000289248	2
RP5-864K19.4	downstream gene variant	630	ENST00000456813	?	?	7
RP5-864K19.4	downstream gene variant	628	ENST00000443161	?	?	7
RP5-864K19.6	downstream gene variant, upstream gene variant	896	ENST00000489803	?	?	4
RP5-864K19.6	downstream gene variant, upstream gene variant	876	ENST00000454994	?	ENSP00000406846	5
RP5-864K19.6	downstream gene variant, upstream gene variant	891	ENST00000489575	?	?	5
RP5-864K19.6	downstream gene variant, upstream gene variant	891	ENST00000621281	?	ENSP00000479064	5
RP5-864K19.7	downstream gene variant, upstream gene variant	116	ENST00000622355	?	?	9
RRAGC	downstream gene variant, upstream gene variant	488	ENST00000373001	NM_022157.3, NM_001271851.1	ENSP00000362092	2
RRAGC	downstream gene variant	2028	ENST00000474456	?	?	1
RRAGC	downstream gene variant, upstream gene variant	2447	ENST00000493015	?	?	3
RRAGC	downstream gene variant, upstream gene variant	642	ENST00000496778	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GJA9	ENST00000357771	NM_030772.4	ENSP00000350415	8
MYCBP	ENST00000465771	?	?	3
MYCBP	ENST00000494695	?	ENSP00000485717	3
MYCBP	ENST00000397572	NM_012333.4	ENSP00000380702	5
MYCBP	ENST00000495043	?	?	3
RHBDL2	ENST00000372990	NM_017821.3, NM_001304746.1	ENSP00000362081	32
RHBDL2	ENST00000289248	?	ENSP00000289248	32
RP5-864K19.4	ENST00000456813	?	?	7
RP5-864K19.4	ENST00000443161	?	?	3
RP5-864K19.4	ENST00000433671	?	?	50
RP5-864K19.6	ENST00000489575	?	?	14
RP5-864K19.6	ENST00000489803	?	?	14
RP5-864K19.6	ENST00000621281	?	ENSP00000479064	14
RP5-864K19.6	ENST00000454994	?	ENSP00000406846	13
RP5-864K19.7	ENST00000622355	?	?	13
RRAGC	ENST00000373001	NM_022157.3, NM_001271851.1	ENSP00000362092	7
RRAGC	ENST00000474456	?	?	6
RRAGC	ENST00000493015	?	?	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RHBDL2	ENST00000372990	NM_017821.3, NM_001304746.1	ENSP00000362081	1

RHBDL2	ENST00000289248	?	ENSP00000289248	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MYCBP	ENST00000397572	NM_012333.4	ENSP00000380702	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP5-864K19.6	ENST00000489803	?	3
RP5-864K19.7	ENST00000622355	?	6
RRAGC	ENST00000474456	?	4