Locus 184 (rs289828)

SNiPAcard

Block annotations

Block info

genomic range	chr2:152,085,984-152,186,147
block size	100,164 bp
variant count	40 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.458 [-3.25 – 1.808]	gene(s) hit or close-by	NMI, RBM43
phastCons	μ = 0.042 [0 – 0.765]	eQTL gene(s)	NMI, RBM43, RIF1, TNFAIP6
GERP++	μ = -0.615 [-5.11 – 2.17]	potentially regulated gene(s)	–
CADD score	μ = 3.579 [0.007 – 10.95]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RIF1	ENST00000453091	ILMN_2149053	blood	3.72×10^{-5} (p-value)	Westra et al.	13
RIF1	ENST00000243326					
NMI	?	ENSG00000123609	testis	8.36×10^{-6} (p-value)	GTEx Portal V6	14
RIF1	?	ENSG00000080345	tibial nerve	5.10×10^{-6} (p-value)	GTEx Portal V6	23
NMI	?	ENSG00000123609	transformed fibroblasts	4.50×10^{-12} (p-value)	GTEx Portal V6	36
TNFAIP6	?	ENSG00000123610	transformed fibroblasts	1.07×10^{-11} (p-value)	GTEx Portal V6	40
RIF1	?	ENSG00000080345	transformed fibroblasts	1.66×10^{-5} (p-value)	GTEx Portal V6	1
RBM43	?	ENSG00000184898	blood	1.35×10^{-9} (p-value)	GTEx Portal V6	40
TNFAIP6	?	ENSG00000123610	blood	1.06×10^{-43} (p-value)	GTEx Portal V6	40
NMI	?	ENSG00000123609	skeletal muscle	6.88×10^{-7} (p-value)	GTEx Portal V6	15
NMI	?	ENSG00000123609	thyroid	7.93×10^{-7} (p-value)	GTEx Portal V6	18
NMI	?	ENSG00000123609	spleen	4.32×10^{-6} (p-value)	GTEx Portal V6	14
NMI	?	ENSG00000123609	lung	1.74×10^{-6} (p-value)	GTEx Portal V6	16
?	?	ILMN_1785732	monocyte	2.96×10^{-64} (p-value)	Zeller et al.	5
RBM43	ENST00000331426	ILMN_1858599	monocyte	1.70×10^{-24} (p-value)	Zeller et al.	5
?	?	ILMN_1785732	monocyte	2.00×10^{-12} (p-value)	Fairfax et al.	3
NMI	ENST00000243346	ILMN_1739541	b-cell	4.61×10^{-4} (p-value)	Fairfax et al.	1
RBM43	ENST00000331426	ILMN_3251415	b-cell	3.44×10^{-5} (p-value)	Fairfax et al.	2
RBM43	ENST00000331426	ILMN_1858599	b-cell	4.64×10^{-14} (p-value)	Fairfax et al.	1
RIF1	?	ENSG00000080345	sun exposed skin	5.51×10^{-7} (p-value)	GTEx Portal V6	7
RIF1	?	ENSG00000080345	unexposed skin	1.57×10^{-5} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000600259 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00001547512 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	DNase1
		Osteobl	H3K27ac
		A549	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00001547513 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K4me2, TAF1, PolII, Rad21, H3K9ac
		HSMMtube	H3K9ac, H3K4me2, H2AZ, DNase1
		blood (K562)	H2AZ, PolII, H3K4me2, DNase1, H3K4me3, Cmyc, H3K9ac, Brg1, H3K27ac, Max
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		blood (GM12878)	Tr4, PolII, Srf, H2AZ, DNase1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K4me3, H3K9ac, H3K4me2, H3K27ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K9ac, H3K27ac, H3K4me3
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, DNase1
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, H3K27ac
ENSR00001547517 (promoter flanking region)	1	liver (HepG2)	H3K27me3
		HSMMtube	H3K27ac, H2AZ, DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H2AZ, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
NMI	downstream gene variant, upstream gene variant	101	ENST00000243346	NM_004688.2	ENSP00000243346	2
NMI	downstream gene variant, upstream gene variant	295	ENST00000491771	?	?	5
NMI	downstream gene variant, upstream gene variant	14	ENST00000477072	?	?	11
NMI	downstream gene variant, upstream gene variant	31	ENST00000414946	?	ENSP00000387373	7
RBM43	downstream gene variant	1099	ENST00000331426	NM_198557.2	ENSP00000331211	1
RBM43	downstream gene variant	876	ENST00000409092	?	ENSP00000386440	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
NMI	ENST00000243346	NM_004688.2	ENSP00000243346	Q	caA/caG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NMI	ENST00000243346	NM_004688.2	ENSP00000243346	23
NMI	ENST00000491771	?	?	19
NMI	ENST00000414946	?	ENSP00000387373	13
NMI	ENST00000477072	?	?	9
RBM43	ENST00000331426	NM_198557.2	ENSP00000331211	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RBM43	ENST00000331426	NM_198557.2	ENSP00000331211	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NMI	ENST00000491771	?	1