Locus 390 (rs4834770)

SNiPAcard

Block annotations

Block info

genomic range	chr4:120,224,565-120,244,029
block size	19,465 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.608 [-3.553 – 0.489]	gene(s) hit or close-by	C4orf3, FABP2
phastCons	μ = 0.074 [0 – 0.763]	eQTL gene(s)	C4orf3, FABP2, KLHL2P1, PDE5A, RP11-33B1.1, RP11-33B1.4, RP11-384K6.2, RP11-384K6.6, RP11-548H18.2, USP53
GERP++	μ = -0.928 [-5.65 – 2.35]	potentially regulated gene(s)	–
CADD score	μ = 2.442 [0.004 – 7.632]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
2-aminobutyrate	8.22×10^{-5}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-33B1.4	?	ENSG00000260091	lung	4.08×10^{-8} (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	lung	9.81×10^{-15} (p-value)	GTEx Portal V6	11
RP11-384K6.6	?	ENSG00000260404	lung	1.65×10^{-6} (p-value)	GTEx Portal V6	10
FABP2	?	ENSG00000145384	atrial appendage	6.63×10^{-8} (p-value)	GTEx Portal V6	1
RP11-33B1.1	?	ENSG00000245958	atrial appendage	4.43×10^{-6} (p-value)	GTEx Portal V6	8
RP11-33B1.1	?	ENSG00000245958	transformed fibroblasts	3.88×10^{-15} (p-value)	GTEx Portal V6	11
RP11-384K6.6	?	ENSG00000260404	transformed fibroblasts	2.68×10^{-6} (p-value)	GTEx Portal V6	10
RP11-33B1.1	?	ENSG00000245958	tibial artery	3.95×10^{-10} (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	breast	4.40×10^{-7} (p-value)	GTEx Portal V6	11
RP11-33B1.4	?	ENSG00000260091	blood	1.09×10^{-8} (p-value)	GTEx Portal V6	11
C4orf3	?	ENSG00000164096	blood	5.54×10^{-9} (p-value)	GTEx Portal V6	11
FABP2	?	ENSG00000145384	blood	7.55×10^{-10} (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	blood	5.93×10^{-7} (p-value)	GTEx Portal V6	9
RP11-384K6.6	?	ENSG00000260404	blood	1.20×10^{-11} (p-value)	GTEx Portal V6	11
RP11-384K6.2	?	ENSG00000225892	blood	1.82×10^{-6} (p-value)	GTEx Portal V6	3
RP11-33B1.1	?	ENSG00000245958	thyroid	9.67×10^{-17} (p-value)	GTEx Portal V6	11

KLHL2P1	?	ENSG00000250412	thyroid	1.88×10-5 (p-value)	GTEx Portal V6	1
RP11-33B1.1	?	ENSG00000245958	skeletal muscle	2.15×10-10 (p-value)	GTEx Portal V6	11
RP11-384K6.6	?	ENSG00000260404	skeletal muscle	1.29×10-6 (p-value)	GTEx Portal V6	10
FABP2	?	ENSG00000145384	transverse colon	1.67×10-9 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	transverse colon	7.12×10-12 (p-value)	GTEx Portal V6	11
RP11-548H18.2	?	ENSG00000249244	EBV lymphocytes	6.00×10-8 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	EBV lymphocytes	7.49×10-9 (p-value)	GTEx Portal V6	11
PDE5A	?	ENSG00000138735	EBV lymphocytes	6.40×10-7 (p-value)	GTEx Portal V6	10
RP11-33B1.1	?	ENSG00000245958	unexposed skin	2.81×10-6 (p-value)	GTEx Portal V6	1
FABP2	?	ENSG00000145384	left ventricle	9.80×10-8 (p-value)	GTEx Portal V6	2
RP11-33B1.1	?	ENSG00000245958	left ventricle	4.44×10-11 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	aorta	1.37×10-7 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	subcutaneous adipocytes	4.04×10-12 (p-value)	GTEx Portal V6	11
RP11-548H18.2	?	ENSG00000249244	subcutaneous adipocytes	3.75×10-6 (p-value)	GTEx Portal V6	10
RP11-33B1.1	?	ENSG00000245958	visceral adipocytes	1.16×10-7 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	cortex	9.60×10-7 (p-value)	GTEx Portal V6	1
RP11-33B1.1	?	ENSG00000245958	nucleus accumbens	2.09×10-8 (p-value)	GTEx Portal V6	11
RP11-548H18.2	?	ENSG00000249244	tibial nerve	5.69×10-10 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	tibial nerve	6.99×10-14 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	sigmoid colon	5.88×10-6 (p-value)	GTEx Portal V6	1
RP11-548H18.2	?	ENSG00000249244	anterior cingulate cortex	9.29×10-9 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	esophagus mucosa	1.03×10-12 (p-value)	GTEx Portal V6	11
FABP2	?	ENSG00000145384	adrenal gland	2.28×10-8 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	adrenal gland	1.44×10-6 (p-value)	GTEx Portal V6	8
RP11-33B1.4	?	ENSG00000260091	testis	3.93×10-10 (p-value)	GTEx Portal V6	11
RP11-33B1.1	?	ENSG00000245958	testis	2.29×10-9 (p-value)	GTEx Portal V6	11
RP11-548H18.2	?	ENSG00000249244	frontal cortex	4.00×10-6 (p-value)	GTEx Portal V6	2
RP11-33B1.1	?	ENSG00000245958	frontal cortex	5.39×10-6 (p-value)	GTEx Portal V6	7
RP11-33B1.4	?	ENSG00000260091	pancreas	6.52×10-6 (p-value)	GTEx Portal V6	7
USP53	ENST00000450251	ILMN_1795275	b-cell	4.35×10-7 (p-value)	Fairfax et al.	1
USP53	ENST00000509769					
USP53	ENST00000274030					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001434619 (promoter flanking region)	3	embryonic stem cell (H1ESC)	Rad21, CTCF
		Osteobl	CTCF, H3K27ac
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	CTCF, Rad21
		A549	H3K27me3, CTCF
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C4orf3	upstream gene variant	2489	ENST00000504110	NM_001001701.3	ENSP00000485697	1
FABP2	downstream gene variant, upstream gene variant	177	ENST00000274024	NM_000134.3	ENSP00000274024	5

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
FABP2	ENST00000274024	NM_000134.3	ENSP00000274024	N	aaT/aaC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C4orf3	ENST00000399075	NM_001170330.1	ENSP00000382026	1
FABP2	ENST00000274024	NM_000134.3	ENSP00000274024	3

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FABP2	ENST00000274024	NM_000134.3	ENSP00000274024	2