

SNiPACard

Block annotations

Block info


genomic range	chr4:120,224,565-120,244,029 <i>e!</i>
block size	19,465 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.608$ [-3.553 – 0.489]	gene(s) hit or close-by	C4orf3 <i>e!</i> , FABP2 <i>e!</i>
phastCons	$\mu = 0.074$ [0 – 0.763]	eQTL gene(s)	C4orf3 <i>e!</i> , FABP2 <i>e!</i> , KLHL2P1 <i>e!</i> , PDE5A <i>e!</i> , RP11-33B1.1 <i>e!</i> , RP11-33B1.4 <i>e!</i> , RP11-384K6.2 <i>e!</i> , RP11-384K6.6 <i>e!</i> , RP11-548H18.2 <i>e!</i> , USP53 <i>e!</i>
GERP++	$\mu = -0.928$ [-5.65 – 2.35]	potentially regulated gene(s)	-
CADD score	$\mu = 2.442$ [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 <i>e!</i>	?	ENSG00000260091 <i>e!</i>	lung	4.08×10 ⁻⁸ (p-value)	GTEEx Portal V6 	11
RP11-33B1.1 <i>e!</i>	?	ENSG00000245958 <i>e!</i>	lung	9.81×10 ⁻¹⁵ (p-value)	GTEEx Portal V6 	11
RP11-384K6.6 <i>e!</i>	?	ENSG00000260404 <i>e!</i>	lung	1.65×10 ⁻⁶ (p-value)	GTEEx Portal V6 	10
FABP2 <i>e!</i>	?	ENSG00000145384 <i>e!</i>	atrial appendage	6.63×10 ⁻⁸ (p-value)	GTEEx Portal V6 	1
RP11-33B1.1 <i>e!</i>	?	ENSG00000245958 <i>e!</i>	atrial appendage	4.43×10 ⁻⁶ (p-value)	GTEEx Portal V6 	8
RP11-33B1.1 <i>e!</i>	?	ENSG00000245958 <i>e!</i>	transformed fibroblasts	3.88×10 ⁻¹⁵ (p-value)	GTEEx Portal V6 	11
RP11-384K6.6 <i>e!</i>	?	ENSG00000260404 <i>e!</i>	transformed fibroblasts	2.68×10 ⁻⁶ (p-value)	GTEEx Portal V6 	10
RP11-33B1.1 <i>e!</i>	?	ENSG00000245958 <i>e!</i>	tibial artery	3.95×10 ⁻¹⁰ (p-value)	GTEEx Portal V6 	11
RP11-33B1.1 <i>e!</i>	?	ENSG00000245958 <i>e!</i>	breast	4.40×10 ⁻⁷ (p-value)	GTEEx Portal V6 	11
RP11-33B1.4 <i>e!</i>	?	ENSG00000260091 <i>e!</i>	blood	1.09×10 ⁻⁸ (p-value)	GTEEx Portal V6 	11
C4orf3 <i>e!</i>	?	ENSG00000164096 <i>e!</i>	blood	5.54×10 ⁻⁹ (p-value)	GTEEx Portal V6 	11
FABP2 <i>e!</i>	?	ENSG00000145384 <i>e!</i>	blood	7.55×10 ⁻¹⁰ (p-value)	GTEEx Portal V6 	11
RP11-33B1.1 <i>e!</i>	?	ENSG00000245958 <i>e!</i>	blood	5.93×10 ⁻⁷ (p-value)	GTEEx Portal V6 	9
RP11-384K6.6 <i>e!</i>	?	ENSG00000260404 <i>e!</i>	blood	1.20×10 ⁻¹¹ (p-value)	GTEEx Portal V6 	11
RP11-384K6.2 <i>e!</i>	?	ENSG00000225892 <i>e!</i>	blood	1.82×10 ⁻⁶ (p-value)	GTEEx Portal V6 	3
RP11-33B1.1 <i>e!</i>	?	ENSG00000245958 <i>e!</i>	throid	9.67×10 ⁻¹⁷ (p-value)	GTEEx Portal V6 	11

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

Putative effect on regulation

Regulatory feature cluster

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

Variation proximal to gene

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

Putative effect on transcript

Synonymous coding variant

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

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C4orf3 <i>e!</i>	ENST00000399075 <i>e!</i>	NM_001170330.1	ENSP00000382026 <i>e!</i>	1
FABP2 <i>e!</i>	ENST00000274024 <i>e!</i>	NM_000134.3	ENSP00000274024 <i>e!</i>	3

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FABP2 <i>e!</i>	ENST00000274024 <i>e!</i>	NM_000134.3	ENSP00000274024 <i>e!</i>	2

