

SNiPACard

Block annotations

Block info

genomic range	chr10:52,017,680-52,029,901 <i>e!</i>
block size	12,222 bp
variant count	22 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.701$ [-3.248 – 2.176]	gene(s) hit or close-by	DYNC112P1 <i>e!</i>
phastCons	$\mu = 0.113$ [0 – 1]	eQTL gene(s)	FAM21A <i>e!</i>
GERP++	$\mu = -0.161$ [-7.14 – 1.41]	potentially regulated gene(s)	-
CADD score	$\mu = 3.195$ [0.039 – 11.92]	disease gene(s)	-

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	muscularis mucosae	4.66×10 ⁻⁶ (p-value)	GTEx Portal V6	14
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	lung	9.80×10 ⁻¹¹ (p-value)	GTEx Portal V6	18
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	atrial appendage	1.18×10 ⁻⁸ (p-value)	GTEx Portal V6	17
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	transformed fibroblasts	2.28×10 ⁻⁹ (p-value)	GTEx Portal V6	16
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	breast	2.34×10 ⁻⁹ (p-value)	GTEx Portal V6	19
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	tibial artery	1.53×10 ⁻¹⁴ (p-value)	GTEx Portal V6	19
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	thyroid	1.61×10 ⁻¹² (p-value)	GTEx Portal V6	19
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	skeletal muscle	3.93×10 ⁻¹⁹ (p-value)	GTEx Portal V6	19
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	liver	1.87×10 ⁻⁹ (p-value)	GTEx Portal V6	17
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	sun exposed skin	1.22×10 ⁻¹⁵ (p-value)	GTEx Portal V6	19
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	unexposed skin	2.21×10 ⁻⁸ (p-value)	GTEx Portal V6	17
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	left ventricle	6.73×10 ⁻⁸ (p-value)	GTEx Portal V6	16
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	subcutaneous adipocytes	2.62×10 ⁻¹⁵ (p-value)	GTEx Portal V6	19
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	visceral adipocytes	5.71×10 ⁻¹³ (p-value)	GTEx Portal V6	19
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	sigmoid colon	3.70×10 ⁻⁷ (p-value)	GTEx Portal V6	17
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	tibial nerve	4.95×10 ⁻¹⁰ (p-value)	GTEx Portal V6	18
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	esophagus mucosa	5.48×10 ⁻¹² (p-value)	GTEx Portal V6	19
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	testis	7.29×10 ⁻⁹ (p-value)	GTEx Portal V6	17
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	putamen	1.59×10 ⁻⁶ (p-value)	GTEx Portal V6	6
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	gastroesophageal junction	7.79×10 ⁻⁷ (p-value)	GTEx Portal V6	8
FAM21A <i>e!</i>	?	ENSG00000099290 <i>e!</i>	hippocampus	2.59×10 ⁻⁶ (p-value)	GTEx Portal V6	4

FAM21A <i>e!</i> ?	ENSG00000099290 <i>e!</i>	frontal cortex	4.18×10 ⁻⁶ (p-value)	GTEx Portal V6 <i>e!</i>	1
FAM21A <i>e!</i> ?	ENSG00000099290 <i>e!</i>	anterior cingulate cortex	1.16×10 ⁻⁶ (p-value)	GTEx Portal V6 <i>e!</i>	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001423388 <i>e!</i> (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		lung (IMR90)	H3K36me3, H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DYNC1I2P1 <i>e!</i>	upstream gene variant, downstream gene variant	77	ENST00000435916 <i>e!</i>	?	?	16

Putative effect on transcript

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
DYNC1I2P1 <i>e!</i>	ENST00000435916 <i>e!</i>	?	3

