Locus 147 (rs4984778)

SNiPAcard

Block annotations

Block info

genomic range	chr16:1,272,472-1,306,040
block size	33,569 bp
variant count	23 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.000 [-3.057 – 0.514]	gene(s) hit or close-by	CACNA1H, PRSS29P, RP11-616M22.12, RP11-616M22.5, TPSAB1, TPSB2, TPSD1, TPSG1
phastCons	μ = 0.004 [0 – 0.019]	eQTL gene(s)	RP11-616M22.3, RP11-616M22.5, TPSAB1, TPSB2, TPSD1, TPSG1
GERP++	μ = -0.650 [-3.81 – 1.1]	potentially regulated gene(s)	CHTF18, TPSD1
CADD score	μ = 2.104 [0.016 – 6.061]	disease gene(s)	CACNA1H

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CACNA1H	EPILEPSY, CHILDHOOD ABSENCE, SUSCEPTIBILITY TO, 6	OMIM	MIM:611942
CACNA1H	Childhood absence epilepsy	OrphaNet	OrphaNet:64280

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TPSD1	?	ENSG00000095917	pancreas	7.27×10-11 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	pancreas	1.36×10-7 (p-value)	GTEx Portal V6	10
TPSD1	?	ENSG00000095917	muscularis mucosae	1.95×10-18 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	muscularis mucosae	2.14×10-19 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	muscularis mucosae	1.74×10-6 (p-value)	GTEx Portal V6	11
TPSD1	?	ENSG00000095917	lung	6.73×10-30 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	lung	1.82×10-26 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	lung	1.88×10-12 (p-value)	GTEx Portal V6	17
TPSAB1	?	ENSG00000172236	blood	2.20×10-7 (p-value)	GTEx Portal V6	10
TPSD1	?	ENSG00000095917	blood	1.98×10-6 (p-value)	GTEx Portal V6	5
TPSD1	?	ENSG00000095917	breast	9.11×10-9 (p-value)	GTEx Portal V6	14
RP11-616M22.5	?	ENSG00000260182	breast	1.46×10-6 (p-value)	GTEx Portal V6	4
TPSB2	?	ENSG00000197253	breast	1.89×10-6 (p-value)	GTEx Portal V6	2
TPSD1	?	ENSG00000095917	tibial artery	1.01×10-17 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	tibial artery	2.69×10-11 (p-value)	GTEx Portal V6	16

TPSD1	?	ENSG00000095917	skeletal muscle	8.05×10-14 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	skeletal muscle	3.31×10-12 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	skeletal muscle	2.00×10-6 (p-value)	GTEx Portal V6	9
TPSD1	?	ENSG00000095917	prostate	1.33×10-12 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	prostate	6.62×10-11 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	putamen	1.27×10-10 (p-value)	GTEx Portal V6	16
TPSD1	?	ENSG00000095917	transverse colon	5.69×10-13 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	transverse colon	5.11×10-10 (p-value)	GTEx Portal V6	15
RP11-616M22.3	?	ENSG00000261294	transverse colon	1.47×10-6 (p-value)	GTEx Portal V6	4
TPSD1	?	ENSG00000095917	sun exposed skin	7.68×10-24 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	sun exposed skin	1.01×10-20 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	sun exposed skin	3.62×10-11 (p-value)	GTEx Portal V6	16
TPSG1	?	ENSG00000116176	sun exposed skin	7.05×10-7 (p-value)	GTEx Portal V6	1
TPSAB1	?	ENSG00000172236	sun exposed skin	5.76×10-6 (p-value)	GTEx Portal V6	1
TPSG1	?	ENSG00000116176	unexposed skin	6.29×10-7 (p-value)	GTEx Portal V6	6
TPSD1	?	ENSG00000095917	unexposed skin	9.92×10-14 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	unexposed skin	7.91×10-13 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	unexposed skin	4.24×10-6 (p-value)	GTEx Portal V6	1
TPSD1	?	ENSG00000095917	aorta	4.69×10-9 (p-value)	GTEx Portal V6	14
TPSB2	?	ENSG00000197253	aorta	4.01×10-7 (p-value)	GTEx Portal V6	7
TPSD1	?	ENSG00000095917	left ventricle	6.33×10-9 (p-value)	GTEx Portal V6	15
TPSD1	?	ENSG00000095917	subcutaneous adipocytes	1.12×10-16 (p-value)	GTEx Portal V6	16
TPSG1	?	ENSG00000116176	subcutaneous adipocytes	7.44×10-10 (p-value)	GTEx Portal V6	17
TPSAB1	?	ENSG00000172236	subcutaneous adipocytes	1.71×10-6 (p-value)	GTEx Portal V6	4
RP11-616M22.5	?	ENSG00000260182	subcutaneous adipocytes	1.07×10-14 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	subcutaneous adipocytes	9.67×10-6 (p-value)	GTEx Portal V6	2
TPSAB1	ENST00000562432	ILMN_1676256	monocyte	2.14×10-21 (p-value)	Zeller et al.	1
TPSAB1	ENST00000561736					
RP11-616M22.5	ENST00000566997					
TPSB2	ENST00000611196					
TPSAB1	ENST00000338844					
TPSB2	ENST00000606293					
TPSAB1	ENST00000461509					
TPSD1	ENST00000211076					
TPSD1	ENST00000397534					
TPSB2	ENST00000612142					
TPSAB1	ENST00000338844	ILMN_2169801	monocyte	1.74×10-19 (p-value)	Zeller et al.	1
TPSAB1	ENST00000461509					
TPSAB1	ENST00000562432					
TPSAB1	ENST00000561736					
TPSB2	?	ENSG00000197253	caudate basal ganglia	1.30×10-15 (p-value)	GTEx Portal V6	17

TPSD1	?	ENSG00000095917	stomach	2.19×10-13 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	stomach	4.06×10-13 (p-value)	GTEx Portal V6	16
TPSD1	?	ENSG00000095917	visceral adipocytes	5.93×10-9 (p-value)	GTEx Portal V6	13
RP11-616M22.5	?	ENSG00000260182	visceral adipocytes	1.96×10-7 (p-value)	GTEx Portal V6	8
TPSB2	?	ENSG00000197253	visceral adipocytes	4.40×10-6 (p-value)	GTEx Portal V6	1
TPSB2	?	ENSG00000197253	nucleus accumbens	4.27×10-12 (p-value)	GTEx Portal V6	15
TPSB2	?	ENSG00000197253	cortex	1.70×10-12 (p-value)	GTEx Portal V6	16
TPSD1	?	ENSG00000095917	tibial nerve	9.35×10-15 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	tibial nerve	7.05×10-11 (p-value)	GTEx Portal V6	16
TPSG1	?	ENSG00000116176	tibial nerve	4.64×10-6 (p-value)	GTEx Portal V6	1
TPSD1	?	ENSG00000095917	esophagus mucosa	1.30×10-22 (p-value)	GTEx Portal V6	17
RP11-616M22.5	?	ENSG00000260182	esophagus mucosa	9.90×10-22 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	esophagus mucosa	1.86×10-11 (p-value)	GTEx Portal V6	17
TPSB2	?	ENSG00000197253	anterior cingulate cortex	3.86×10-12 (p-value)	GTEx Portal V6	15
TPSD1	?	ENSG00000095917	gastroesophageal junction	6.16×10-11 (p-value)	GTEx Portal V6	16
RP11-616M22.5	?	ENSG00000260182	gastroesophageal junction	1.33×10-9 (p-value)	GTEx Portal V6	13
TPSD1	?	ENSG00000095917	testis	6.86×10-9 (p-value)	GTEx Portal V6	14
RP11-616M22.5	?	ENSG00000260182	testis	2.96×10-10 (p-value)	GTEx Portal V6	16
TPSB2	?	ENSG00000197253	frontal cortex	1.13×10-12 (p-value)	GTEx Portal V6	15
TPSD1	?	ENSG00000095917	atrial appendage	2.56×10-7 (p-value)	GTEx Portal V6	10
RP11-616M22.5	?	ENSG00000260182	atrial appendage	1.76×10-7 (p-value)	GTEx Portal V6	11
TPSB2	?	ENSG00000197253	hypothalamus	4.64×10-10 (p-value)	GTEx Portal V6	14
TPSB2	?	ENSG00000197253	hippocampus	2.27×10-9 (p-value)	GTEx Portal V6	9
TPSD1	?	ENSG00000095917	terminal ileum	1.33×10-6 (p-value)	GTEx Portal V6	1
RP11-616M22.5	?	ENSG00000260182	terminal ileum	3.45×10-7 (p-value)	GTEx Portal V6	1
RP11-616M22.3	?	ENSG00000261294	terminal ileum	2.33×10-7 (p-value)	GTEx Portal V6	2
TPSD1	?	ENSG00000095917	thyroid	9.03×10-7 (p-value)	GTEx Portal V6	8
RP11-616M22.5	?	ENSG00000260182	cerebellum	2.30×10-6 (p-value)	GTEx Portal V6	3
TPSG1	ENST00000564684	ILMN_1769219	skin	4.76×10-8 (p-value)	MuTHER consortium	1
TPSG1	ENST00000234798		adipocyte	2.12×10-5 (p-value)	MuTHER consortium	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000020937		

ENCODE promoter-associated distal DHS (Enhancer)

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000184452	1	ENCP00000020927	CHTF18

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000502225 (open chromatin region)	4	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	DNase1, H3K27me3
		A549	H3K27me3, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CACNA1H	downstream gene variant	704	ENST00000348261	NM_021098.2	ENSP00000334198	4
CACNA1H	downstream gene variant	1388	ENST00000562079	?	ENSP00000454581	1
CACNA1H	downstream gene variant	1388	ENST00000569107	?	ENSP00000454990	1
CACNA1H	downstream gene variant	1388	ENST00000565831	?	ENSP00000455840	1
CACNA1H	downstream gene variant	707	ENST00000564927	?	ENSP00000455512	4
CACNA1H	downstream gene variant	1388	ENST00000564231	?	ENSP00000457555	1
CACNA1H	downstream gene variant	701	ENST00000358590	NM_001005407.1	ENSP00000351401	4
PRSS29P	downstream gene variant	4913	ENST00000568091	?	?	1
RP11-616M22.12	upstream gene variant, downstream gene variant	1167	ENST00000621827	?	?	6
RP11-616M22.5	upstream gene variant	1300	ENST00000566997	?	?	4
TPSAB1	upstream gene variant, downstream gene variant	1027	ENST00000561736	?	ENSP00000456821	9
TPSAB1	upstream gene variant, downstream gene variant	500	ENST00000461509	?	ENSP00000418247	10
TPSAB1	upstream gene variant, downstream gene variant	383	ENST00000562432	?	?	9
TPSAB1	upstream gene variant, downstream gene variant	472	ENST00000338844	NM_003294.3	ENSP00000343577	10
TPSB2	downstream gene variant, upstream gene variant	274	ENST00000612142	?	ENSP00000478695	6
TPSB2	downstream gene variant, upstream gene variant	542	ENST00000611196	?	ENSP00000484461	5
TPSB2	downstream gene variant, upstream gene variant	273	ENST00000606293	NM_024164.5	ENSP00000482743	6
TPSD1	upstream gene variant	20	ENST00000397534	?	ENSP00000380668	4
TPSD1	upstream gene variant	94	ENST00000211076	NM_012217.2	ENSP00000211076	4
TPSG1	downstream gene variant, upstream gene variant	187	ENST00000564684	?	?	6
TPSG1	upstream gene variant	1151	ENST00000234798	NM_012467.3	ENSP00000234798	5

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
TPSB2	ENST00000611196	?	ENSP00000484461	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TPSB2	ENST00000611196	?	ENSP00000484461	1

TPSG1	ENST00000234798	NM_012467.3	ENSP00000234798	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TPSAB1	ENST00000338844	NM_003294.3	ENSP00000343577	1
TPSAB1	ENST00000461509	?	ENSP00000418247	1
TPSAB1	ENST00000561736	?	ENSP00000456821	1