Locus 322 (rs2729354)

SNiPAcard

Block annotations

Block info

genomic range	chr11:57,283,988-57,358,343
block size	74,356 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.373 [-5.515 – 0.209]	**gene(s) hit or close-by**	RPS4XP13, SLC43A1, SMTNL1, UBE2L6
phastCons	μ = 0.025 [0 – 0.132]	**eQTL gene(s)**	BTBD18, RN7SL605P, RP11-624G17.3, SERPING1, SLC43A1, SMTNL1, TIMM10, YPEL4, ZDHHC5
GERP++	μ = -0.744 [-6.2 – 1.34]	**potentially regulated gene(s)**	CLP1, PRG2, RP11-872D17.8, RTN4RL2, SLC43A1, SLC43A3, YPEL4, ZDHHC5
CADD score	μ = 2.266 [0.264 – 4.946]	**disease gene(s)**	SERPING1, CLP1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SERPING1	ANGIOEDEMA, HEREDITARY, TYPE I	OMIM	MIM:106100
CLP1	PONTOCEREBELLAR HYPOPLASIA, TYPE 10	OMIM	MIM:615803
CLP1	Pontocerebellar hypoplasia, type 10	DECIPHER	MIM:615803
SERPING1	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147
SERPING1	Hereditary angioedema type 1	OrphaNet	OrphaNet:100050
SERPING1	Hereditary angioedema type 2	OrphaNet	OrphaNet:100051

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TIMM10	?	ENSG00000134809	atrial appendage	1.07×10^{-24} (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	atrial appendage	1.54×10^{-6} (p-value)	GTEx Portal V6	9
TIMM10	?	ENSG00000134809	transformed fibroblasts	3.08×10^{-47} (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	transformed fibroblasts	3.76×10^{-11} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	blood	1.60×10^{-8} (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	breast	1.91×10^{-19} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	skeletal muscle	2.01×10^{-29} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	putamen	9.09×10^{-13} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	EBV lymphocytes	4.67×10^{-27} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	ovary	3.38×10^{-12} (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	sun exposed skin	3.82×10^{-24} (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	sun exposed skin	2.11×10^{-7} (p-value)	GTEx Portal V6	16

BTBD18	?	ENSG00000233436	unexposed skin	1.93×10-5 (p-value)	GTEx Portal V6	1
TIMM10	?	ENSG00000134809	unexposed skin	3.89×10-25 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	aorta	5.62×10-36 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	aorta	7.26×10-6 (p-value)	GTEx Portal V6	2
TIMM10	?	ENSG00000134809	left ventricle	5.09×10-26 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	left ventricle	1.42×10-5 (p-value)	GTEx Portal V6	3
TIMM10	?	ENSG00000134809	subcutaneous adipocytes	1.03×10-29 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	subcutaneous adipocytes	1.90×10-8 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	caudate basal ganglia	6.91×10-9 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	visceral adipocytes	1.54×10-23 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	stomach	5.18×10-23 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	cortex	3.61×10-9 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	tibial nerve	4.16×10-25 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	tibial nerve	1.71×10-10 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	esophagus mucosa	8.37×10-55 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	anterior cingulate cortex	1.85×10-10 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	pituitary	3.78×10-10 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	testis	1.16×10-30 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	testis	4.08×10-6 (p-value)	GTEx Portal V6	4
TIMM10	?	ENSG00000134809	hypothalamus	5.83×10-8 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	uterus	3.71×10-9 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	pancreas	5.35×10-20 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	muscularis mucosae	1.59×10-43 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	muscularis mucosae	7.49×10-6 (p-value)	GTEx Portal V6	3
TIMM10	?	ENSG00000134809	lung	6.47×10-19 (p-value)	GTEx Portal V6	16
YPEL4	?	ENSG00000166793	lung	3.64×10-7 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	lung	2.00×10-10 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	tibial artery	1.08×10-40 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	tibial artery	1.51×10-8 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	thyroid	1.02×10-31 (p-value)	GTEx Portal V6	16
SMTNL1	?	ENSG00000214872	thyroid	8.44×10-7 (p-value)	GTEx Portal V6	13
TIMM10	?	ENSG00000134809	prostate	2.26×10-11 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	transverse colon	3.33×10-30 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	liver	4.85×10-11 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	terminal ileum	1.23×10-8 (p-value)	GTEx Portal V6	15

TIMM10	?	ENSG00000134809	spleen	1.69×10-8 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	nucleus accumbens	2.59×10-9 (p-value)	GTEx Portal V6	15
SLC43A1	?	ENSG00000149150	nucleus accumbens	1.18×10-7 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	sigmoid colon	4.44×10-20 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	coronary artery	3.72×10-15 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	vagina	7.78×10-7 (p-value)	GTEx Portal V6	3
TIMM10	?	ENSG00000134809	gastroesophageal junction	3.99×10-26 (p-value)	GTEx Portal V6	16
TIMM10	?	ENSG00000134809	adrenal gland	8.70×10-17 (p-value)	GTEx Portal V6	16
SLC43A1	?	ENSG00000149150	adrenal gland	3.43×10-9 (p-value)	GTEx Portal V6	15
TIMM10	?	ENSG00000134809	hippocampus	2.94×10-10 (p-value)	GTEx Portal V6	16
TIMM10	ENST00000525587	ILMN_1765332	skin	8.47×10-79 (p-value)	MuTHER consortium	6
RN7SL605P	ENST00000577229		blood	7.64×10-69 (p-value)	MuTHER consortium	6
TIMM10	ENST00000257245		adipocyte	2.20×10-97 (p-value)	MuTHER consortium	6
TIMM10	?	ENSG00000134809	brain	5.71×10-8 (q-value)	SeeQTL DB (Myers et al.)	3
TIMM10	?	ENSG00000134809	blood	3.36×10-6 (q-value)	SeeQTL DB (HapMap)	4
TIMM10	ENST00000525587	ILMN_1765332	monocyte	<1.0×10-298 (p-value)	Zeller et al.	4
RN7SL605P	ENST00000577229					
TIMM10	ENST00000257245					
YPEL4	ENST00000532314	ILMN_1726624	monocyte	3.05×10-16 (p-value)	Zeller et al.	4
YPEL4	ENST00000528797					
YPEL4	ENST00000300022					
YPEL4	ENST00000524669					
YPEL4	ENST00000524592					
SERPING1	ENST00000278407	ILMN_1670305	blood	1.14×10-6 (p-value)	Westra et al.	5
SERPING1	ENST00000528996					
SERPING1	ENST00000531797					
SERPING1	ENST00000403558					
SERPING1	ENST00000530113					
SERPING1	ENST00000619430					
SERPING1	ENST00000378324					
SERPING1	ENST00000340687					
SERPING1	ENST00000531133					
SERPING1	ENST00000378323					
ZDHHC5	ENST00000529480	ILMN_1679358	blood	3.36×10-6 (p-value)	Westra et al.	6
ZDHHC5	ENST00000287169					
SMTNL1	?	ENSG00000214872	frontal cortex	3.97×10-6 (p-value)	GTEx Portal V6	9
SMTNL1	?	ENSG00000214872	cerebellar hemisphere	3.21×10-6 (p-value)	GTEx Portal V6	12
RP11-624G17.3	?	ENSG00000255301	cerebellar hemisphere	6.56×10-6 (p-value)	GTEx Portal V6	1
SLC43A1	?	ENSG00000149150	cerebellum	9.33×10-6 (p-value)	GTEx Portal V6	8
ZDHHC5	ENST00000529480	ILMN_1679358	b-cell	2.23×10-5 (p-value)	Fairfax et al.	2
ZDHHC5	ENST00000287169					
YPEL4	ENST00000528797	ILMN_1726624	monocyte	2.62×10-4 (p-value)	Fairfax et al.	2

YPEL4	ENST00000532314					
YPEL4	ENST00000524669					
YPEL4	ENST00000300022					
YPEL4	ENST00000524592					
RN7SL605P	ENST00000577229	ILMN_1765332	monocyte	1.02×10-63 (p-value)	Fairfax et al.	2
TIMM10	ENST00000525587		b-cell	5.82×10-46 (p-value)	Fairfax et al.	2
TIMM10	ENST00000257245					
RN7SL605P	ENST00000577229	ILMN_1765332	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
TIMM10	ENST00000525587					
TIMM10	ENST00000257245					
TIMM10	ENST00000525158	1555764_s_at	blood	1.50×10-21 (p-value)	Dixon et al.	1
TIMM10	ENST00000525587					
TIMM10	ENST00000257245					
TIMM10	ENST00000525158	218408_at	blood	2.30×10-17 (p-value)	Dixon et al.	1
TIMM10	ENST00000525587					
TIMM10	ENST00000257245					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
TIMM10	?	ENSG00000134809	chr11	brain	1.29×10-5 (q-value)	SeeQTL DB (Myers et al.)	3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000106365	1	ENCP00000012246	SLC43A3
		ENCP00000012254	SLC43A1
		ENCP00000012242	SLC43A3
		ENCP00000012255	SLC43A1
		ENCP00000012278	ZDHHC5
		ENCP00000012240	PRG2
		ENCP00000012241	RP11-872D17.8
		ENCP00000012247	SLC43A3
		ENCP00000012244	RP11-872D17.8
		ENCP00000012256	SLC43A1
		ENCP00000012276	CLP1
		ENCP00000012275	YPEL4
		ENCP00000012249	RTN4RL2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000319083 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (K562)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPS4XP13	upstream gene variant	2321	ENST00000483219	?	?	1
SLC43A1	upstream gene variant	1682	ENST00000534105	?	ENSP00000432241	1
SLC43A1	upstream gene variant	1664	ENST00000530159	?	ENSP00000434312	1
SLC43A1	upstream gene variant	735	ENST00000524995	?	ENSP00000432361	1

SLC43A1	upstream gene variant	735	ENST00000524995	?	ENSP00000432361	1
SLC43A1	upstream gene variant	729	ENST00000278426	NM_003627.5	ENSP00000278426	1
SLC43A1	upstream gene variant	1630	ENST00000528450	NM_001198810.1	ENSP00000435673	1
SLC43A1	upstream gene variant	1900	ENST00000533066	?	ENSP00000435647	1
SLC43A1	upstream gene variant	829	ENST00000533263	?	ENSP00000435486	1
SMTNL1	downstream gene variant	3575	ENST00000527972	NM_001105565.2	ENSP00000432651	1
SMTNL1	downstream gene variant	3575	ENST00000399154	?	ENSP00000382108	1
UBE2L6	upstream gene variant	2661	ENST00000528275	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
UBE2L6	ENST00000528275	?	?	4
UBE2L6	ENST00000526659	?	ENSP00000434348	5
UBE2L6	ENST00000287156	NM_004223.4	ENSP00000287156	5
UBE2L6	ENST00000527022	?	ENSP00000435265	5
UBE2L6	ENST00000340573	NM_198183.2	ENSP00000341980	5