Locus 29 (rs11603020)

SNiPAcard

Block annotations

Block info

genomic range	chr11:57,369,008-57,400,462
block size	31,455 bp
variant count	32 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.465 [-3.014 – 1.413]	gene(s) hit or close-by	SERPING1
phastCons	μ = 0.024 [0 – 0.267]	eQTL gene(s)	AP000662.4, MED19, RN7SL605P, TIMM10, YPEL4, ZDHHC5
GERP++	μ = -0.478 [-6.6 – 3.68]	potentially regulated gene(s)	CLP1, PRG2, RP11-872D17.8, RTN4RL2, SERPING1, SERPING1, SLC43A1, SLC43A3, SSRP1, YPEL4, ZDHHC5
CADD score	μ = 3.907 [0.037 – 22.6]	disease gene(s)	SERPING1, CLP1

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	not provided	ClinVar	RCV000077812.1	1

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
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
CLP1	Pontocerebellar hypoplasia, type 10	DECIPHER	MIM:615803

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
SERPING1	missense variant	ENST00000340687	?	ENSP00000341861	M/V	Atg/Gtg	?	?	1
SERPING1	missense variant	ENST00000278407	NM_001032295.1, NM_000062.2	ENSP00000278407	M/V	Atg/Gtg	?	?	1
SERPING1	missense variant	ENST00000403558	?	ENSP00000384420	M/V	Atg/Gtg	?	?	1
SERPING1	missense variant	ENST00000378324	?	ENSP00000367575	M/V	Atg/Gtg	?	?	1
SERPING1	missense variant	ENST00000378323	?	ENSP00000367574	M/V	Atg/Gtg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TIMM10	?	ENSG00000134809	muscularis mucosae	2.46×10^{-10} (p-value)	GTEx Portal V6	32

TIMM10	?	ENSG00000134809	lung	7.44×10-9 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	tibial nerve	1.51×10-9 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	thyroid	1.81×10-7 (p-value)	GTEx Portal V6	32
AP000662.4	?	ENSG00000254602	thyroid	2.01×10-10 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	esophagus mucosa	1.45×10-11 (p-value)	GTEx Portal V6	32
MED19	?	ENSG00000156603	esophagus mucosa	3.39×10-6 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	skeletal muscle	7.74×10-6 (p-value)	GTEx Portal V6	8
ZDHHC5	?	ENSG00000156599	skeletal muscle	8.30×10-6 (p-value)	GTEx Portal V6	12
TIMM10	?	ENSG00000134809	sun exposed skin	1.11×10-7 (p-value)	GTEx Portal V6	32
MED19	?	ENSG00000156603	sun exposed skin	2.16×10-9 (p-value)	GTEx Portal V6	32
TIMM10	?	ENSG00000134809	subcutaneous adipocytes	7.61×10-7 (p-value)	GTEx Portal V6	24
MED19	?	ENSG00000156603	subcutaneous adipocytes	7.87×10-7 (p-value)	GTEx Portal V6	32
RN7SL605P	ENST00000577229	ILMN_1765332	monocyte	6.00×10-5 (p-value)	Fairfax et al.	5
TIMM10	ENST00000525587		b-cell	1.31×10-4 (p-value)	Fairfax et al.	5
TIMM10	ENST00000257245					
TIMM10	ENST00000525587	ILMN_1765332	skin	3.74×10-7 (p-value)	MuTHER consortium	7
RN7SL605P	ENST00000577229		blood	9.92×10-7 (p-value)	MuTHER consortium	7
TIMM10	ENST00000257245		adipocyte	6.66×10-10 (p-value)	MuTHER consortium	7
TIMM10	ENST00000525587	ILMN_1765332	monocyte	1.71×10-25 (p-value)	Zeller et al.	3
RN7SL605P	ENST00000577229					
TIMM10	ENST00000257245					
YPEL4	ENST00000532314	ILMN_1726624	monocyte	4.38×10-21 (p-value)	Zeller et al.	3
YPEL4	ENST00000528797					
YPEL4	ENST00000300022					
YPEL4	ENST00000524669					
YPEL4	ENST00000524592					
TIMM10	?	ENSG00000134809	stomach	8.05×10-6 (p-value)	GTEx Portal V6	8
TIMM10	?	ENSG00000134809	pancreas	1.55×10-5 (p-value)	GTEx Portal V6	2

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000066147	1	ENST00000403558, ENST00000278407, ENST00000378324, ENST00000340687, ENST00000378323	SERPING1
FFCP00000066188	1	ENST00000403558, ENST00000530113, ENST00000278407, ENST00000378324, ENST00000340687, ENST00000378323	SERPING1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000106368	1	ENCP00000012246	SLC43A3
		ENCP00000012254	SLC43A1
		ENCP00000012242	SLC43A3

		ENCP00000012278	ZDHHC5
		ENCP00000012241	RP11-872D17.8
		ENCP00000012240	PRG2
		ENCP00000012247	SLC43A3
		ENCP00000012244	RP11-872D17.8
		ENCP00000012256	SLC43A1
		ENCP00000012276	CLP1
		ENCP00000012275	YPEL4
		ENCP00000012249	RTN4RL2
ENCE00000106266	2	ENCP00000012246	SLC43A3
		ENCP00000012268	SERPING1 SERPING1
		ENCP00000012254	SLC43A1
		ENCP00000012242	SLC43A3
		ENCP00000012255	SLC43A1
		ENCP00000012278	ZDHHC5
		ENCP00000012235	SSRP1
		ENCP00000012241	RP11-872D17.8
		ENCP00000012240	PRG2
		ENCP00000012247	SLC43A3
		ENCP00000012244	RP11-872D17.8
		ENCP00000012256	SLC43A1
		ENCP00000012276	CLP1
		ENCP00000012275	YPEL4
		ENCP00000012249	RTN4RL2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001570401 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		lung (IMR90)	H3K36me3
		blood (K562)	H3K27me3
ENSR00001603770 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K36me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SERPING1	downstream gene variant	1090	ENST00000403558	?	ENSP00000384420	4
SERPING1	upstream gene variant, downstream gene variant	505	ENST00000531605	?	?	8
SERPING1	downstream gene variant	1323	ENST00000531133	?	ENSP00000435431	4
SERPING1	downstream gene variant	1227	ENST00000405496	?	ENSP00000384561	10
SERPING1	downstream gene variant	1057	ENST00000378324	?	ENSP00000367575	4
SERPING1	upstream gene variant, downstream gene variant	1071	ENST00000528996	?	ENSP00000431226	14
SERPING1	downstream gene variant	1443	ENST00000457869	?	ENSP00000399746	10
SERPING1	upstream gene variant, downstream gene variant	736	ENST00000530113	?	?	10
SERPING1	downstream gene variant	1323	ENST00000619430	?	ENSP00000478572	4
SERPING1	downstream gene variant	1051	ENST00000278407	NM_001032295.1, NM_000062.2	ENSP00000278407	4
SERPING1	downstream gene variant	1058	ENST00000340687	?	ENSP00000341861	4

SERPING1	downstream gene variant	1114	ENST00000531797	?		ENSP00000432554	4
SERPING1	downstream gene variant	1165	ENST00000378323	?		ENSP00000367574	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPING1	ENST00000531133	?	ENSP00000435431	17
SERPING1	ENST00000531797	?	ENSP00000432554	17
SERPING1	ENST00000278407	NM_001032295.1, NM_000062.2	ENSP00000278407	17
SERPING1	ENST00000403558	?	ENSP00000384420	17
SERPING1	ENST00000528996	?	ENSP00000431226	7
SERPING1	ENST00000619430	?	ENSP00000478572	17
SERPING1	ENST00000378323	?	ENSP00000367574	17
SERPING1	ENST00000378324	?	ENSP00000367575	17
SERPING1	ENST00000531605	?	?	8
SERPING1	ENST00000340687	?	ENSP00000341861	17
SERPING1	ENST00000530113	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPING1	ENST00000531133	?	ENSP00000435431	1
SERPING1	ENST00000531797	?	ENSP00000432554	1
SERPING1	ENST00000528996	?	ENSP00000431226	1
SERPING1	ENST00000619430	?	ENSP00000478572	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SERPING1	ENST00000530113	?	1