

SNiPACard

Block annotations

Block info

genomic range	chr14:95,022,686-95,040,431 <i>e!</i>
block size	17,746 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.617$ [-2.753 – 1.541]	gene(s) hit or close-by	SERPINA4 <i>e!</i> , SERPINA5 <i>e!</i>
phastCons	$\mu = 0.049$ [0 – 0.979]	eQTL gene(s)	SERPINA3 <i>e!</i> , SERPINA3 <i>e!</i> , SERPINA5 <i>e!</i>
GERP++	$\mu = -1.273$ [-6.99 – 2.58]	potentially regulated gene(s)	-
CADD score	$\mu = 2.503$ [0.079 – 9.785]	disease gene(s)	SERPINA3 <i>e!</i>

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SERPINA3 <i>e!</i>	Alpha-1-antichymotrypsin deficiency	OrphaNet	OrphaNet:93594 orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SERPINA5 <i>e!</i>	?	ENSG00000188488 <i>e!</i>	tibial artery	1.44×10 ⁻⁹ (p-value)	GTEx Portal V6 M	20
SERPINA3 <i>e!</i>	ENST00000467132 <i>e!</i>	ILMN_1788874 <i>e!</i>	adipocyte	2.28×10 ⁻¹³ (p-value)	MuTHER consortium M	15
SERPINA3 <i>e!</i>	ENST00000393080 <i>e!</i>					
SERPINA3 <i>e!</i>	ENST00000556968 <i>e!</i>					
SERPINA3 <i>e!</i>	ENST00000553947 <i>e!</i>					
SERPINA3 <i>e!</i>	ENST00000482740 <i>e!</i>					
SERPINA3 <i>e!</i>	ENST00000393078 <i>e!</i>					
SERPINA5 <i>e!</i>	?	ENSG00000188488 <i>e!</i>	skeletal muscle	1.75×10 ⁻⁸ (p-value)	GTEx Portal V6 M	10
SERPINA5 <i>e!</i>	?	ENSG00000188488 <i>e!</i>	thyroid	1.90×10 ⁻⁶ (p-value)	GTEx Portal V6 M	6

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001459927 <i>e!</i> (enhancer)	3	liver (HepG2) Osteobl blood (DND-41)	H3K27ac, H3K4me2, H3K4me1 H2AZ, H3K27ac H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SERPINA4 <i>e!</i>	upstream gene variant, downstream gene variant	57	ENST00000555095 <i>e!</i>	NM_001289032.1	ENSP00000451172 <i>e!</i>	8
SERPINA4 <i>e!</i>	upstream gene variant, downstream gene variant	840	ENST00000557004 <i>e!</i>	NM_006215.3	ENSP00000450838 <i>e!</i>	8
SERPINA4 <i>e!</i>	upstream gene variant, downstream gene variant	62	ENST00000298841 <i>e!</i>	NM_001289033.1	ENSP00000298841 <i>e!</i>	8

SERPINA5 <i>e!</i>	upstream gene variant	57	ENST00000554220 <i>e!</i> ?	ENSP00000450484 <i>e!</i>	3
SERPINA5 <i>e!</i>	upstream gene variant	57	ENST00000553780 <i>e!</i> ?	ENSP00000450837 <i>e!</i>	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
SERPINA4 <i>e!</i>	ENST00000555095 <i>e!</i>	NM_001289032.1	ENSP00000451172 <i>e!</i>	F	ttT/ttC	1
SERPINA4 <i>e!</i>	ENST00000557004 <i>e!</i>	NM_006215.3	ENSP00000450838 <i>e!</i>	F	ttT/ttC	1
SERPINA4 <i>e!</i>	ENST00000298841 <i>e!</i>	NM_001289033.1	ENSP00000298841 <i>e!</i>	F	ttT/ttC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPINA4 <i>e!</i>	ENST00000555095 <i>e!</i>	NM_001289032.1	ENSP00000451172 <i>e!</i>	12
SERPINA4 <i>e!</i>	ENST00000557004 <i>e!</i>	NM_006215.3	ENSP00000450838 <i>e!</i>	12
SERPINA4 <i>e!</i>	ENST00000298841 <i>e!</i>	NM_001289033.1	ENSP00000298841 <i>e!</i>	12
SERPINA5 <i>e!</i>	ENST00000554220 <i>e!</i>	?	ENSP00000450484 <i>e!</i>	17
SERPINA5 <i>e!</i>	ENST00000553780 <i>e!</i>	?	ENSP00000450837 <i>e!</i>	17

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPINA4 <i>e!</i>	ENST00000557004 <i>e!</i>	NM_006215.3	ENSP00000450838 <i>e!</i>	1

