Locus 81 (rs882732)

SNiPAcard

Block annotations

Block info

genomic range	chr14:95,022,686-95,040,431
block size	17,746 bp
variant count	21 variants

Basic features

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

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SERPINA3	Alpha-1-antichymotrypsin deficiency	OrphaNet	OrphaNet:93594

Direct effect on regulation

cis-eQTL

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

Putative effect on regulation

Regulatory feature cluster

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

Variation proximal to gene

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

SERPINA5	upstream gene variant	57	ENST00000554220	?	ENSP00000450484	3
SERPINA5	upstream gene variant	57	ENST00000553780	?	ENSP00000450837	3

Putative effect on transcript

Synonymous coding variant

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

Intron variant

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

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPINA4	ENST00000557004	NM_006215.3	ENSP00000450838	1