Locus 320 (rs4905179)

SNiPAcard

Block annotations

Block info

genomic range	chr14:94,784,618-94,818,078
block size	33,461 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.179 [-0.31 – 1.321]	gene(s) hit or close-by	SERPINA6
phastCons	μ = 0.005 [0 – 0.021]	eQTL gene(s)	SERPINA1
GERP++	μ = -0.556 [-4.11 – 1.31]	potentially regulated gene(s)	–
CADD score	μ = 2.992 [0.741 – 8.487]	disease gene(s)	SERPINA6, SERPINA1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Breast size	<8.00×10-7	GWAS Catalog	22747683	1

Disease gene annotation

gene	trait	source DB	source entry/link
SERPINA6	CORTICOSTEROID-BINDING GLOBULIN DEFICIENCY	OMIM	MIM:611489
SERPINA1	ALPHA-1-ANTITRYPSIN DEFICIENCY	OMIM	MIM:613490
SERPINA6	CORTICOSTEROID-BINDING GLOBULIN DEFICIENCY	OrphaNet	OrphaNet:199247
SERPINA1	ALPHA-1-ANTITRYPSIN DEFICIENCY	OrphaNet	OrphaNet:60
SERPINA1	Hemorrhagic disease due to alpha-1 antitrypsin Pittsburgh mutation	OrphaNet	OrphaNet:178396

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SERPINA1	ENST00000393087	ILMN_1764980	blood	2.42×10-5 (p-value)	Westra et al.	2
SERPINA1	ENST00000557361					
SERPINA1	ENST00000554720					
SERPINA1	ENST00000557093					
SERPINA1	ENST00000556091					
SERPINA1	ENST00000393087	ILMN_2256050	monocyte	5.08×10-15 (p-value)	Zeller et al.	1
SERPINA1	ENST00000557361					
SERPINA1	ENST00000555289					
SERPINA1	ENST00000557093					
SERPINA1	ENST00000393088					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000652586	1	liver (HepG2)	H3K4me3, H3K27ac, H3K9ac, H3K4me2, H3K4me1, H3K79me2, PolII

(promoter flanking region)		HSMMtube	H3K27me3
		A549	DNase1
		blood (DND-41)	H3K27me3
		skin (NHEK)	DNase1
ENSR00000422985 (CTCF binding site)	1	liver (HepG2)	CTCF
		HSMMtube	H3K27me3
ENSR00000422992 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	H3K4me1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K27me3
		liver (HepG2)	H3K4me2, H3K4me1, Jund, FOSL2, DNase1
		blood (GM12878)	H3K27ac, NFKB, DNase1, H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K27me3
		skin (NHEK)	H3K27me3, H3K4me1, DNase1
ENSR00001624850 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me1, H3K27ac
		cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	H3K4me2, H3K4me1
		blood (GM12878)	H3K4me1, DNase1, NFKB, H3K27ac
		A549	H3K27me3
		skin (NHEK)	H3K27me3, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SERPINA6	upstream gene variant	2714	ENST00000341584	NM_001756.3	ENSP00000342850	1
SERPINA6	upstream gene variant	2789	ENST00000555056	?	ENSP00000451045	1
SERPINA6	upstream gene variant	2719	ENST00000557225	?	ENSP00000452018	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SERPINA6	ENST00000341584	NM_001756.3	ENSP00000342850	2
SERPINA6	ENST00000555056	?	ENSP00000451045	2
SERPINA6	ENST00000557225	?	ENSP00000452018	2