Locus 282 (rs2749534)

SNiPAcard

Block annotations

Block info

genomic range	chr14:94,809,760-94,841,641
block size	31,882 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.175 [-4.333 – 0.735]	gene(s) hit or close-by	SERPINA1, SERPINA2
phastCons	μ = 0.016 [0 – 0.343]	eQTL gene(s)	–
GERP++	μ = -0.914 [-4.49 – 2.08]	potentially regulated gene(s)	SERPINA1, SERPINA1, SERPINA9
CADD score	μ = 2.358 [0.063 – 7.213]	disease gene(s)	SERPINA1

Trait annotations

Disease gene annotation

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

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)
SERPINA2	start lost	ENST00000553483	?	ENSP00000486888	I/M	atA/atG	?	benign	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000172474	1	ENCP00000019874	SERPINA1 SERPINA1
ENCE00000172475	1	ENCP00000019874	SERPINA1 SERPINA1
ENCE00000172476	1	ENCP00000019874	SERPINA1 SERPINA1
		ENCP00000019876	SERPINA9

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001459896 (promoter flanking region)	1	NHLF	DNase1, H3K27ac
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K4me1, H3K27ac, DNase1
		breast (HMEC)	H3K4me2, H3K27ac, DNase1

		monocytes (Monocytes-CD14+)	DNase1
		liver (HepG2)	H3K27ac, H3K4me2, H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1, H3K4me2
		A549	DNase1, H3K27me3
		skin (NHEK)	DNase1
ENSR00001624849 (enhancer)	2	cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	H3K4me1
ENSR00000422992 (promoter flanking region)	3	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
ENSR00001459901 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	Rad21, Nrsf, CTCF, Yy1, DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac
		blood (K562)	CTCF, H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K4me1, H3K27ac
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K27me3
		liver (HepG2)	CTCF, H3K4me1, Rad21, PolII
		lung (IMR90)	DNase1
		A549	CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SERPINA1	downstream gene variant	1443	ENST00000437397	NM_001127706.1, NM_001127702.1, NM_001127707.1	ENSP00000408474	2
SERPINA1	downstream gene variant	1443	ENST00000440909	?	ENSP00000390299	2
SERPINA1	downstream gene variant	1443	ENST00000448921	NM_001127705.1, NM_001127701.1, NM_001002236.2, NM_001127704.1, NM_001127703.1	ENSP00000416066	2
SERPINA1	downstream gene variant	1443	ENST00000355814	NM_001127700.1, NM_001002235.2	ENSP00000348068	2
SERPINA1	downstream gene variant	3145	ENST00000449399	?	ENSP00000416354	1
SERPINA1	downstream gene variant	3085	ENST00000404814	?	ENSP00000385960	1
SERPINA1	downstream gene variant	3072	ENST00000393088	?	ENSP00000376803	1
SERPINA1	downstream gene variant	3045	ENST00000393087	NM_000295.4	ENSP00000376802	1
SERPINA1	downstream gene variant	3809	ENST00000402629	?	ENSP00000386094	1
SERPINA1	downstream gene variant	3069	ENST00000489769	?	ENSP00000451525	1
SERPINA2	downstream gene variant, upstream gene variant	53	ENST00000553483	?	ENSP00000486888	8