Locus 270 (rs6575449)

SNiPAcard

Block annotations

Block info

genomic range	chr14:95,094,014-95,097,666
block size	3,653 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.513 [-1.412 – -0.013]	gene(s) hit or close-by	ADIPOR1P2, SERPINA3, SERPINA3
phastCons	μ = 0.001 [0 – 0.002]	eQTL gene(s)	–
GERP++	μ = -0.490 [-2.45 – 1.3]	potentially regulated gene(s)	–
CADD score	μ = 2.221 [0.102 – 6.546]	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)
?	?	ILMN_1838581	monocyte	5.48×10-4 (p-value)	Fairfax et al.	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADIPOR1P2	upstream gene variant	2376	ENST00000554199	?	?	6
SERPINA3	downstream gene variant	3622	ENST00000393080	?	ENSP00000376795	1
SERPINA3	downstream gene variant	3625	ENST00000393078	NM_001085.4	ENSP00000376793	1
SERPINA3	downstream gene variant	3623	ENST00000556968	?	ENSP00000452476	1
SERPINA3	downstream gene variant	3625	ENST00000482740	?	ENSP00000451119	1
SERPINA3	downstream gene variant	3623	ENST00000467132	?	ENSP00000450540	1
SERPINA3	downstream gene variant	3031	ENST00000553947	?	ENSP00000452367	2
SERPINA3	downstream gene variant	4914	ENST00000556388	?	?	1