SNiPAcard

Block annotations

Block info

genomic range	chr11:18,277,763-18,287,750
block size	9,988 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.167 [-2.326 – -0.307]	**gene(s) hit or close-by**	RNA5SP334, SAA1, ST13P5
phastCons	μ = 0.001 [0 – 0.004]	**eQTL gene(s)**	GTF2H1, SAA1, SAA2, SAA2-SAA4
GERP++	μ = -1.923 [-6.95 – 1.81]	**potentially regulated gene(s)**	HPS5, PTPN5, SAA1, TSG101
CADD score	μ = 1.247 [0.123 – 3.047]	**disease gene(s)**	SAA1, HPS5

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Amyloid A serum levels	$<3.00\times10^{-53}$	GWAS Catalog	21124955	1

Disease gene annotation

gene	trait	source DB	source entry/link
SAA1	Secondary amyloidosis	OrphaNet	OrphaNet:85445
HPS5	Hermansky-Pudlak syndrome without pulmonary fibrosis	OrphaNet	OrphaNet:231512
HPS5	HERMANSKY-PUDLAK SYNDROME 5	OMIM	MIM:614074

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SAA1	?	ENSG00000173432	tibial artery	6.31×10^{-7} (p-value)	GTEx Portal V6	6
SAA2	?	ENSG00000134339	tibial artery	3.39×10^{-6} (p-value)	GTEx Portal V6	6
SAA2-SAA4	?	ENSG00000255071	tibial artery	8.31×10^{-6} (p-value)	GTEx Portal V6	6
GTF2H1	ENST00000534641	ILMN_2157957	blood	7.08×10^{-6} (p-value)	Westra et al.	4
GTF2H1	ENST00000265963					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000055282	1	ENST00000356524, ENST00000532858, ENST00000405158	SAA1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000100066	1	ENCP00000011406	HPS5
		ENCP00000011423	TSG101
		ENCP00000011443	PTPN5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000559405 (enhancer)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	H3K4me1
		Osteobl	H3K27me3
		blood (GM12878)	H3K4me1
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00001568722 (enhancer)	2	NHLF	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		breast (HMEC)	H3K4me1
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RNA5SP334	upstream gene variant	190	ENST00000364825	?	?	3
SAA1	upstream gene variant	687	ENST00000356524	NM_199161.3	ENSP00000348918	2
SAA1	upstream gene variant	61	ENST00000405158	NM_000331.4	ENSP00000384906	3
SAA1	upstream gene variant	57	ENST00000532858	NM_001178006.1	ENSP00000436866	3
ST13P5	upstream gene variant, downstream gene variant	1136	ENST00000526214	?	?	6

Putative effect on transcript

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SAA1	ENST00000356524	NM_199161.3	ENSP00000348918	1