Locus 243 (rs4938330)

SNiPAcard

Block annotations

Block info

genomic range	chr11:116,779,499-116,982,821
block size	203,323 bp
variant count	63 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.780 [-4.61 – 2.46]	gene(s) hit or close-by	AP000936.1, AP000936.2, AP000936.4, RNY4P6, SIK3
phastCons	μ = 0.041 [0 – 1]	eQTL gene(s)	APOA1, APOA1-AS, BUD13, PCSK7, RP11-109L13.1, SIDT2, SIK3, TAGLN
GERP++	μ = -0.556 [-5.77 – 3.97]	potentially regulated gene(s)	AP000936.1, BACE1
CADD score	μ = 3.543 [0.006 – 19.14]	disease gene(s)	APOA1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Cholesterol, hdl	9.34×10^{-5}	dbGaP	pha003074	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
APOA1	AMYLOIDOSIS, FAMILIAL VISCERAL	OMIM	MIM:105200	OMIM
APOA1	HYPOALPHALIPOPROTEINEMIA, PRIMARY	OMIM	MIM:604091	OMIM
APOA1	TANGIER DISEASE	OMIM	MIM:205400	OMIM
APOA1	APOLIPOPROTEIN A-I	OMIM	MIM:107680	OMIM
APOA1	Primary systemic amyloidosis	OrphaNet	OrphaNet:314701	orphanet
APOA1	Familial renal amyloidosis due to Apolipoprotein AI variant	OrphaNet	OrphaNet:93560	orphanet
APOA1	APOLIPOPROTEIN A-I DEFICIENCY	OrphaNet	OrphaNet:425	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TAGLN	ENST00000525531	ILMN_1778668	blood	3.34×10^{-8} (p-value)	MuTHER consortium	26
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
RP11-109L13.1	?	ENSG00000254851	visceral adipocytes	7.68×10^{-11} (p-value)	GTEx Portal V6	61
RP11-109L13.1	?	ENSG00000254851	testis	1.98×10^{-7} (p-value)	GTEx Portal V6	59
APOA1	?	ENSG00000118137	left ventricle	5.60×10^{-12} (p-value)	GTEx Portal V6	62
APOA1-AS	?	ENSG00000235910	left ventricle	8.11×10^{-11} (p-value)	GTEx Portal V6	62
RP11-109L13.1	?	ENSG00000254851	thyroid	6.71×10^{-9} (p-value)	GTEx Portal V6	59

SIK3	?	ENSG00000160584	esophagus mucosa	4.60×10-9 (p-value)	GTEx Portal V6	59
PCSK7	?	ENSG00000160613	esophagus mucosa	6.36×10-8 (p-value)	GTEx Portal V6	28
RP11-109L13.1	?	ENSG00000254851	esophagus mucosa	4.73×10-6 (p-value)	GTEx Portal V6	14
RP11-109L13.1	?	ENSG00000254851	muscularis mucosae	7.17×10-7 (p-value)	GTEx Portal V6	17
RP11-109L13.1	?	ENSG00000254851	stomach	8.58×10-8 (p-value)	GTEx Portal V6	42
RP11-109L13.1	?	ENSG00000254851	tibial nerve	3.96×10-8 (p-value)	GTEx Portal V6	21
PCSK7	?	ENSG00000160613	transformed fibroblasts	9.62×10-7 (p-value)	GTEx Portal V6	39
TAGLN	?	ENSG00000149591	blood	6.52×10-12 (p-value)	GTEx Portal V6	60
BUD13	?	ENSG00000137656	blood	5.63×10-6 (p-value)	GTEx Portal V6	4
RP11-109L13.1	?	ENSG00000254851	tibial artery	9.61×10-8 (p-value)	GTEx Portal V6	34
TAGLN	?	ENSG00000149591	EBV lymphocytes	1.81×10-7 (p-value)	GTEx Portal V6	8
RP11-109L13.1	?	ENSG00000254851	sun exposed skin	3.14×10-8 (p-value)	GTEx Portal V6	21
RP11-109L13.1	?	ENSG00000254851	aorta	2.03×10-7 (p-value)	GTEx Portal V6	20
RP11-109L13.1	?	ENSG00000254851	subcutaneous adipocytes	3.42×10-9 (p-value)	GTEx Portal V6	23
RP11-109L13.1	?	ENSG00000254851	atrial appendage	6.58×10-6 (p-value)	GTEx Portal V6	1
RP11-109L13.1	?	ENSG00000254851	unexposed skin	3.78×10-6 (p-value)	GTEx Portal V6	14
SIDT2	ENST00000278951	ILMN_1791912	blood	9.93×10-7 (p-value)	Westra et al.	25
SIDT2	ENST00000532062					
SIDT2	ENST00000324225					
SIDT2	ENST00000431081					
SIDT2	ENST00000620360					
RP11-109L13.1	?	ENSG00000254851	lung	7.78×10-6 (p-value)	GTEx Portal V6	12
RP11-109L13.1	?	ENSG00000254851	skeletal muscle	9.17×10-6 (p-value)	GTEx Portal V6	7
TAGLN	ENST00000525531	ILMN_1778668	monocyte	9.43×10-14 (p-value)	Zeller et al.	2
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000014771		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000123692	1	ENCP00000014802	BACE1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001573920 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me3, H3K27ac, DNase1, H3K4me1
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K4me2
		Osteobl	H3K27ac
		muscle (HSMM)	DNase1
ENSR00001573922 (promoter flanking region)	1	NHLF	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27ac
		liver (HepG2)	H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00001607331 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H4K20me1
		blood (GM12878)	H3K4me2, H3K4me1
		A549	H3K4me2, H3K4me3
ENSR00000569539 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		embryonic stem cell (H1ESC)	DNase1
		blood (K562)	DNase1
		blood (DND-41)	H3K4me1, H3K27ac
ENSR00001607333 (enhancer)	1	monocytes (Monocytes-CD14+)	H4K20me1
		Osteobl	H3K36me3
ENSR00000569542 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H4K20me1
		endothelium (HUVEC)	Cjun, DNase1
		nervous (NH-A)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000569548 (promoter flanking region)	8	embryonic stem cell (H1ESC)	DNase1, CTCF
		HSMMtube	DNase1
		blood (K562)	PU1, Cjun, FOSL1, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		liver (HepG2)	FOSL2, Jund, DNase1, H3K4me1, FOXA1
		lung (IMR90)	DNase1
		blood (GM12878)	H3K79me2, BATF
		nervous (NH-A)	DNase1, H3K27me3
		skin (NHEK)	H3K27ac, H3K36me3, H3K4me1, DNase1
		NHLF	DNase1
		Osteobl	H3K27ac, H4K20me1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1, Cfos, Jund, Cjun
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H4K20me1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001607339 (CTCF binding site)	2	monocytes (Monocytes-CD14+)	H3K4me1, H4K20me1
		liver (HepG2)	H3K4me1
		A549	H3K4me2, H3K4me3
		skin (NHEK)	H3K4me1, H3K27ac
ENSR00001607340 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H4K20me1
		embryonic stem cell (H1ESC)	CTCF
		blood (GM12878)	H3K79me2
		nervous (NH-A)	H3K27me3
		skin (NHEK)	DNase1
ENSR00001573930 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H4K20me1
		embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H4K20me1, FOXA2, HDAC2, DNase1, H3K4me1, FOXA1
		blood (GM12878)	H3K4me2, H3K79me2, H3K4me1
		skin (NHEK)	DNase1
		muscle (HSMM)	H3K36me3
ENSR00001573932	1	monocytes (Monocytes-CD14+)	H4K20me1

(enhancer)			
ENSR00001573933 (enhancer)	1	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, FOXA1
		blood (GM12878)	H3K36me3, H3K4me2, H3K4me3, H3K79me2
		A549	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP000936.1	downstream gene variant, upstream gene variant	276	ENST00000438127	?	?	5
AP000936.2	downstream gene variant, upstream gene variant	50	ENST00000584092	?	?	2
AP000936.4	downstream gene variant	1025	ENST00000442124	?	?	2
RNY4P6	upstream gene variant, downstream gene variant	517	ENST00000363667	?	?	2
SIK3	upstream gene variant	361	ENST00000413553	?	ENSP00000414347	2
SIK3	upstream gene variant	341	ENST00000497049	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SIK3	ENST00000497049	?	?	4
SIK3	ENST00000446921	NM_001281749.1	ENSP00000390442	61
SIK3	ENST00000485363	?	?	61
SIK3	ENST00000413553	?	ENSP00000414347	4
SIK3	ENST00000445177	?	ENSP00000391295	61
SIK3	ENST00000415541	?	ENSP00000392761	61
SIK3	ENST00000375300	NM_001281748.1, NM_025164.4	ENSP00000364449	61

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AP000936.1	ENST00000438127	?	1