Locus 150 (rs12099358)

SNiPAcard

Block annotations

Block info

genomic range	chr11:116,718,415-116,972,935
block size	254,521 bp
variant count	77 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.215 [-3.588 – 2.455]	**gene(s) hit or close-by**	AP000936.1, AP000936.2, AP000936.4, APOA1-AS, RNY4P6, SIK3, SIK3-IT1
phastCons	μ = 0.049 [0 – 0.909]	**eQTL gene(s)**	APOA1, APOA1-AS, SIDT2
GERP++	μ = -0.360 [-8.15 – 3.25]	**potentially regulated gene(s)**	AP000936.4, BUD13, CEP164, PCSK7, RNF214, SIDT2, SIK3
CADD score	μ = 4.261 [0.03 – 18.86]	**disease gene(s)**	APOA1, CEP164

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
2-palmitoylglycerophosphocholine*	2.41×10-5	Metabolomics GWAS Server	24816252	5
1-oleoylglycerol (1-monoolein)	5.16×10-5	Metabolomics GWAS Server	24816252	4

Disease gene annotation

gene	trait	source DB	source entry/link
APOA1	AMYLOIDOSIS, FAMILIAL VISCERAL	OMIM	MIM:105200
APOA1	HYPOALPHALIPOPROTEINEMIA, PRIMARY	OMIM	MIM:604091
APOA1	TANGIER DISEASE	OMIM	MIM:205400
APOA1	APOLIPOPROTEIN A-I	OMIM	MIM:107680
CEP164	NEPHRONOPHTHISIS 15	OMIM	MIM:614845
APOA1	Primary systemic amyloidosis	OrphaNet	OrphaNet:314701
APOA1	Familial renal amyloidosis due to Apolipoprotein AI variant	OrphaNet	OrphaNet:93560
APOA1	APOLIPOPROTEIN A-I DEFICIENCY	OrphaNet	OrphaNet:425
CEP164	Senior-Loken syndrome	OrphaNet	OrphaNet:3156

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
APOA1	?	ENSG00000118137	left ventricle	6.90×10-11 (p-value)	GTEx Portal V6	76
APOA1-AS	?	ENSG00000235910	left ventricle	1.34×10-9 (p-value)	GTEx Portal V6	74
SIDT2	ENST00000278951	ILMN_1791912	blood	8.62×10-5 (p-value)	Westra et al.	9
SIDT2	ENST00000532062					
SIDT2	ENST00000324225					
SIDT2	ENST00000431081					
SIDT2	ENST00000620360					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000123650	1	ENCP00000014795	PCSK7
ENCE00000123336	1	ENCP00000014749	BUD13
		ENCP00000014780	SIDT2
ENCE00000123493	1	ENCP00000014770	SIK3
ENCE00000123628	1	ENCP00000014793	PCSK7
ENCE00000123403	1	ENCP00000014798	PCSK7 RNF214
		ENCP00000014772	SIK3 AP000936.4
		ENCP00000014804	CEP164

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000569505 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K36me3
		blood (GM12878)	PU1, H3K4me3, EBF1, DNase1, Yy1, H3K4me1, H3K27ac, H3K36me3, H3K4me2
		lung (IMR90)	H3K36me3, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1, H3K36me3
		NHLF	DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3
		breast (HMEC)	H3K4me1, H3K27ac
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K36me3
		endothelium (HUVEC)	H3K4me1, DNase1
		A549	H3K36me3
ENSR00001573904 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K9ac, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1, H3K4me1
		muscle (HSMM)	H3K4me1, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K4me2, H3K36me3
		NHLF	H3K36me3, DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K36me3, H3K4me1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		A549	H3K36me3
ENSR00000190303 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K4me2
		blood (DND-41)	H3K4me1, H3K36me3
		skin (NHDF-AD)	H3K4me3
		breast (HMEC)	H3K4me2
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K36me3

		liver (HepG2)	H3K4me1, H3K4me2, HNF4A
		blood (GM12878)	H3K4me1
		lung (IMR90)	H3K4me2
		skin (NHEK)	H3K4me2
ENSR00000569523 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K4me3, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00001573919 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1
		nervous (NH-A)	DNase1
ENSR00000321726 (enhancer)	1	blood (K562)	DNase1
ENSR00000569533 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, USF1
		HSMMtube	DNase1, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K4me1
		NHLF	DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		breast (HMEC)	H3K4me1, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Jund, Cjun
		monocytes (Monocytes-CD14+)	DNase1, H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	DNase1
ENSR00001607329 (enhancer)	1	liver (HepG2)	H3K4me1
ENSR00001573924 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	Jund
		monocytes (Monocytes-CD14+)	H4K20me1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me2, H3K4me3
		skin (NHEK)	DNase1
ENSR00000569539 (enhancer)	2	monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		embryonic stem cell (H1ESC)	DNase1
		blood (K562)	DNase1
		blood (DND-41)	H3K4me1, H3K27ac
ENSR00000569540 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H4K20me1, H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		skin (NHEK)	H3K4me1, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1
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)	2	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
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
ENSR00000569552 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H4K20me1, H3K79me2
		blood (GM12878)	H3K79me2
		skin (NHEK)	DNase1, H3K4me1, H3K4me2
		breast (HMEC)	H3K4me2, H3K9ac, H3K27ac, DNase1
ENSR00001573933 (enhancer)	2	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
ENSR00000569556 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, CTCF, PolII, Rad21, TAF7, Sin3Ak20, H3K27ac, ATF3, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3
		HSMMtube	DNase1, H3K9ac, H3K4me2, H3K27ac, H2AZ
		blood (K562)	USF1, H3K79me2, H2AZ, TAF1, Max, PolII, H3K4me2, CTCF, DNase1, H3K4me3, H3K9ac, Rad21, H3K27me3, Yy1, H3K27ac
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K9ac, DNase1
		liver (HepG2)	Yy1, H3K27me3, H3K4me3, CTCF, H3K79me2, Rad21, TAF1, USF1, H3K4me1, H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, DNase1
		lung (IMR90)	CTCF, H3K9ac, H3K4me3, H3K4me2, H3K27ac, DNase1
		blood (GM12878)	Yy1, DNase1, H3K4me1, H2AZ, PolII, PU1, ATF3, Rad21, H3K79me2, H3K9ac, H3K4me2, H3K27ac, USF1, H3K4me3
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K4me2, H3K4me3, DNase1, H3K9ac, H3K27ac, CTCF
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	CTCF, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me3, H3K27ac, DNase1, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K4me3, PolII, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, DNase1
		A549	H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP000936.1	downstream gene variant, upstream gene variant	2526	ENST00000438127	?	?	3

AP000936.2	downstream gene variant, upstream gene variant	2369	ENST00000584092	?	?	4
AP000936.4	upstream gene variant	905	ENST00000442124	?	?	2
APOA1-AS	downstream gene variant	1020	ENST00000444200	?	?	1
RNY4P6	upstream gene variant, downstream gene variant	2836	ENST00000363667	?	?	4
SIK3	upstream gene variant	4152	ENST00000413553	?	ENSP00000414347	1
SIK3	downstream gene variant, upstream gene variant	738	ENST00000472648	?	?	3
SIK3	downstream gene variant, upstream gene variant	4016	ENST00000497049	?	?	2
SIK3	downstream gene variant	165	ENST00000480468	?	?	3
SIK3	upstream gene variant	3883	ENST00000454905	?	ENSP00000392849	1
SIK3	upstream gene variant	3824	ENST00000445177	?	ENSP00000391295	1
SIK3	upstream gene variant	586	ENST00000488337	?	?	2
SIK3	upstream gene variant	3914	ENST00000485363	?	?	1
SIK3	upstream gene variant	3798	ENST00000375300	NM_001281748.1, NM_025164.4	ENSP00000364449	1
SIK3	upstream gene variant	3782	ENST00000446921	NM_001281749.1	ENSP00000390442	1
SIK3	upstream gene variant	3948	ENST00000415541	?	ENSP00000392761	1
SIK3-IT1	downstream gene variant, upstream gene variant	1178	ENST00000454832	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AP000936.4	ENST00000442124	?	?	1
APOA1-AS	ENST00000444200	?	?	4
SIK3	ENST00000497049	?	?	18
SIK3	ENST00000465421	?	?	5
SIK3	ENST00000446921	NM_001281749.1	ENSP00000390442	76
SIK3	ENST00000480222	?	?	2
SIK3	ENST00000445177	?	ENSP00000391295	76
SIK3	ENST00000454905	?	ENSP00000392849	2
SIK3	ENST00000415541	?	ENSP00000392761	76
SIK3	ENST00000413553	?	ENSP00000414347	23
SIK3	ENST00000488337	?	?	5
SIK3	ENST00000375300	NM_001281748.1, NM_025164.4	ENSP00000364449	76
SIK3	ENST00000485363	?	?	63
SIK3-IT1	ENST00000454832	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
APOA1-AS	ENST00000444200	?	1