Locus 54 (rs5167)

SNiPAcard

Block annotations

Block info

genomic range	chr19:45,440,529-45,448,465
block size	7,937 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.913 [-2.819 – 1.155]	**gene(s) hit or close-by**	APOC2, APOC4, APOC4-APOC2, CTB-129P6.11
phastCons	μ = 0.016 [0 – 0.043]	**eQTL gene(s)**	APOC1, APOE, CLPTM1
GERP++	μ = 0.266 [-0.38 – 2.07]	**potentially regulated gene(s)**	BCL3, CEACAM19
CADD score	μ = 2.814 [0.159 – 13.66]	**disease gene(s)**	APOE, APOC2

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM065970	1

Disease gene annotation

gene	trait	source DB	source entry/link
APOE	SEA-BLUE HISTIOCYTE DISEASE	OMIM	MIM:269600
APOE	ALZHEIMER DISEASE 2	OMIM	MIM:104310
APOE	HYPERCHOLESTEROLEMIA, FAMILIAL	OMIM	MIM:143890
APOE	LIPOPROTEIN GLOMERULOPATHY	OMIM	MIM:611771
APOE	APOLIPOPROTEIN E	OMIM	MIM:107741
APOC2	APOLIPOPROTEIN C-II DEFICIENCY	OMIM	MIM:207750
APOE	Hyperlipoproteinemia type 3	OrphaNet	OrphaNet:412
APOE	Sea-blue histiocytosis	OrphaNet	OrphaNet:158029
APOE	LIPOPROTEIN GLOMERULOPATHY	OrphaNet	OrphaNet:329481
APOC2	Familial apolipoprotein C-II deficiency	OrphaNet	OrphaNet:309020

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)
APOC4	missense variant	ENST00000592954	NM_001646.2	ENSP00000468236	R/L	cGc/cTc	?	?	1
APOC4-APOC2	missense variant	ENST00000585685	?	ENSP00000467185	R/L	cGc/cTc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CLPTM1	?	ENSG00000104853	testis	1.50×10^{-10} (p-value)	GTEx Portal V6	9
APOE	?	ENSG00000130203	lung	3.76×10^{-8} (p-value)	GTEx Portal V6	8

APOC1	?	ENSG00000130208	lung	7.37×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000230231	1	ENCP00000024489	BCL3
		ENCP00000024485	CEACAM19

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
APOC2	upstream gene variant	827	ENST00000592257	?	ENSP00000477261	4
APOC2	upstream gene variant	848	ENST00000591597	?	ENSP00000476835	4
APOC2	upstream gene variant	3078	ENST00000585786	?	ENSP00000465001	1
APOC2	upstream gene variant	1995	ENST00000590360	?	ENSP00000466775	2
APOC4	upstream gene variant, downstream gene variant	192	ENST00000591600	?	ENSP00000466444	10
APOC4	upstream gene variant	849	ENST00000592954	NM_001646.2	ENSP00000468236	10
APOC4-APOC2	upstream gene variant	774	ENST00000252490	NM_000483.4	ENSP00000252490	4
APOC4-APOC2	upstream gene variant	867	ENST00000589057	?	ENSP00000468139	10
APOC4-APOC2	upstream gene variant	849	ENST00000585685	?	ENSP00000467185	10
CTB-129P6.11	downstream gene variant	4836	ENST00000591646	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOC4	ENST00000591600	?	ENSP00000466444	1
APOC4	ENST00000592954	NM_001646.2	ENSP00000468236	1
APOC4-APOC2	ENST00000585685	?	ENSP00000467185	1
APOC4-APOC2	ENST00000589057	?	ENSP00000468139	2