SNiPAcard

Block annotations

Block info

genomic range	chr2:21,239,862-21,262,427
block size	22,566 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.553 [-4.111 – 6.554]	**gene(s) hit or close-by**	AC010872.1, APOB
phastCons	μ = 0.250 [0 – 1]	**eQTL gene(s)**	–
GERP++	μ = 0.025 [-4.21 – 5.73]	**potentially regulated gene(s)**	–
CADD score	μ = 5.580 [0.276 – 18.42]	**disease gene(s)**	APOB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Lipoproteins, ldl	9.04×10-5	dbGaP	pha002902	dbGaP	1

Variant annotation

trait	type	source DB	source entry/link		Variant(s)
Clinvar: phenotype not specified	likely benign	ClinVar	RCV000116384.2	ClinVar	1
?	HGMD curated	HGMD	CM984191	HGMD®	1

Disease gene annotation

gene	trait	source DB	source entry/link	
APOB	Homozygous familial hypercholesterolemia	OrphaNet	OrphaNet:391665	orphanet
APOB	Familial hypobetalipoproteinemia	OrphaNet	OrphaNet:426	orphanet

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)
APOB	missense variant	ENST00000616098	?	ENSP00000477990	V/A	gTt/gCt	?	?	1
APOB	missense variant	ENST00000233242	NM_000384.2	ENSP00000233242	V/A	gTt/gCt	?	?	1
APOB	missense variant	ENST00000399256	?	ENSP00000382200	V/A	gTt/gCt	?	?	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC010872.1	downstream gene variant	3872	ENST00000624225	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
APOB	ENST00000616098	?	ENSP00000477990	3
APOB	ENST00000233242	NM_000384.2	ENSP00000233242	3
APOB	ENST00000399256	?	ENSP00000382200	2