Locus 71 (rs646776)

SNiPAcard

Block annotations

Block info

genomic range	chr1:109,817,192-109,822,166
block size	4,975 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.610 [-1.832 – 0.394]	**gene(s) hit or close-by**	CELSR2, PSRC1
phastCons	μ = 0.102 [0 – 0.992]	**eQTL gene(s)**	CELSR2, PSRC1, SORT1
GERP++	μ = -1.693 [-9 – 3.35]	**potentially regulated gene(s)**	AMPD2
CADD score	μ = 4.646 [0.153 – 13.51]	**disease gene(s)**	SORT1, AMPD2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Coronary Artery Disease	$<1.00\times10^{-8}$	GWAS Catalog	24262325	1
Coronary artery disease or large artery stroke	$<8.00\times10^{-17}$	GWAS Catalog	24262325	1
Coronary artery disease or ischemic stroke	$<1.00\times10^{-8}$	GWAS Catalog	24262325	1
Cholesterol, total	$<2.00\times10^{-170}$	GWAS Catalog	24097068	1
LDL cholesterol	$<5.00\times10^{-241}$	GWAS Catalog	24097068	1
Lipoprotein-associated phospholipase A2 activity and mass	$<2.00\times10^{-22}$	GWAS Catalog	23118302	1
Lipoprotein-associated phospholipase A2 activity and mass	$<1.00\times10^{-17}$	GWAS Catalog	22003152	1
Cardiovascular disease risk factors	$<2.00\times10^{-22}$	GWAS Catalog	21943158	1
Metabolite levels	$<2.00\times10^{-19}$	GWAS Catalog	21909109	1
Coronary heart disease	$<3.00\times10^{-10}$	GWAS Catalog	21378990	1
Coronary heart disease	$<6.00\times10^{-10}$	GWAS Catalog	21378988	1
LDL cholesterol	$<9.00\times10^{-29}$	GWAS Catalog	21347282	1
Coronary Artery Disease	$<8.00\times10^{-11}$	GWAS Catalog	21239051	1
Progranulin levels	$<2.00\times10^{-30}$	GWAS Catalog	21087763	1
LDL cholesterol	$<1.00\times10^{-26}$	GWAS Catalog	20864672	1
Cholesterol, total	$<6.00\times10^{-131}$	GWAS Catalog	20686565	1
LDL cholesterol	$<1.00\times10^{-170}$	GWAS Catalog	20686565	1
Lipoprotein-associated phospholipase A2 activity and mass	$<3.00\times10^{-15}$	GWAS Catalog	20442857	1
Response to statin therapy	$<4.00\times10^{-6}$	GWAS Catalog	20339536	1
Lipid metabolism phenotypes	$<2.00\times10^{-53}$	GWAS Catalog	19936222	1
Myocardial infarction (early onset)	$<8.00\times10^{-12}$	GWAS Catalog	19198609	1
Cholesterol, total	$<9.00\times10^{-22}$	GWAS Catalog	19060911	1
LDL cholesterol	$<8.00\times10^{-23}$	GWAS Catalog	19060911	1
LDL cholesterol	$<2.00\times10^{-12}$	GWAS Catalog	19060910	1
LDL cholesterol	$<2.00\times10^{-42}$	GWAS Catalog	19060906	1
LDL cholesterol	$<1.00\times10^{-33}$	GWAS Catalog	18262040	1

LDL cholesterol	$<3.00\times10^{-29}$	GWAS Catalog	18193044	1
LDL cholesterol	$<6.00\times10^{-33}$	GWAS Catalog	18193043	1
LDL cholesterol	$<1.00\times10^{-7}$	GWAS Catalog	18179892	1
Coronary heart disease	$<4.00\times10^{-9}$	GWAS Catalog	17634449	1
cholesterol	5.02×10^{-9}	Metabolomics GWAS Server	24816252	7
Cholesterol	2.68×10^{-5}	dbGaP	pha003078	1
Cholesterol	7.39×10^{-5}	dbGaP	pha003073	1
Lipoproteins, ldl	2.19×10^{-12}	dbGaP	pha002902	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Low density lipoprotein cholesterol level quantitative trait locus 6	association	ClinVar	RCV000007497.1	1
?	HGMD curated	HGMD	CR114612	1
?	HGMD curated	HGMD	CR105220	1
LOW DENSITY LIPOPROTEIN CHOLESTEROL LEVEL QUANTITATIVE TRAIT LOCUS 6	OMIM curated	OMIM	MIM:602458	1

Disease gene annotation

gene	trait	source DB	source entry/link
SORT1	LOW DENSITY LIPOPROTEIN CHOLESTEROL LEVEL QUANTITATIVE TRAIT [...]	OMIM	MIM:613589
AMPD2	PONTOCEREBELLAR HYPOPLASIA, TYPE 9	OMIM	MIM:615809
AMPD2	SPASTIC PARAPLEGIA 63, AUTOSOMAL RECESSIVE	OMIM	MIM:615686
AMPD2	Autosomal recessive spastic paraplegia type 63	OrphaNet	OrphaNet:401805
AMPD2	Pontocerebellar hypoplasia, type 9	OrphaNet	OrphaNet:369920

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PSRC1	?	ENSG00000134222	pancreas	2.83×10^{-7} (p-value)	GTEx Portal V6	10
SORT1	?	ENSG00000134243	pancreas	2.33×10^{-6} (p-value)	GTEx Portal V6	5
PSRC1	?	ENSG00000134222	blood	1.98×10^{-7} (p-value)	GTEx Portal V6	5
PSRC1	?	ENSG00000134222	esophagus mucosa	5.45×10^{-12} (p-value)	GTEx Portal V6	10
CELSR2	?	ENSG00000143126	esophagus mucosa	4.25×10^{-7} (p-value)	GTEx Portal V6	7
PSRC1	ENST00000369907	ILMN_1671843	monocyte	6.65×10^{-18} (p-value)	Fairfax et al.	4
PSRC1	ENST00000369909					
PSRC1	ENST00000369904					
PSRC1	ENST00000409138					
PSRC1	ENST00000409267					
PSRC1	ENST00000492431					
PSRC1	ENST00000369903					
CELSR2	?	ENSG00000143126	skeletal muscle	7.42×10^{-36} (p-value)	GTEx Portal V6	10
PSRC1	?	ENSG00000134222	testis	3.79×10^{-8} (p-value)	GTEx Portal V6	10
SORT1	?	ENSG00000134243	liver	3.42×10^{-27} (p-value)	GTEx Portal V6	10
PSRC1	?	ENSG00000134222	liver	4.39×10^{-19} (p-value)	GTEx Portal V6	10

CELSR2	?	ENSG00000143126	liver	3.42×10-19 (p-value)	GTEx Portal V6	10
CELSR2	?	ENSG00000143126	sun exposed skin	7.33×10-6 (p-value)	GTEx Portal V6	2
CELSR2	ENST00000498157	ILMN_1711208	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
CELSR2	ENST00000271332					
SORT1	ENST00000538502	ILMN_1707077	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
SORT1	ENST00000256637					
PSRC1	?	ENSG00000134222	cortex	8.68×10-7 (p-value)	GTEx Portal V6	6
PSRC1	?	ENSG00000134222	unexposed skin	2.73×10-6 (p-value)	GTEx Portal V6	3
PSRC1	ENST00000369907	ILMN_1671843	monocyte	7.21×10-61 (p-value)	Zeller et al.	2
PSRC1	ENST00000369909					
PSRC1	ENST00000369904					
PSRC1	ENST00000409138					
PSRC1	ENST00000409267					
PSRC1	ENST00000369903					
PSRC1	ENST00000492431					
PSRC1	ENST00000369909	ILMN_2315964	monocyte	3.05×10-13 (p-value)	Zeller et al.	2
PSRC1	ENST00000369904					
PSRC1	ENST00000409267					
PSRC1	ENST00000369907					
PSRC1	ENST00000409138					
PSRC1	ENST00000474126					
PSRC1	ENST00000492431					
PSRC1	ENST00000369903					
PSRC1	?	ENSG00000134222	thyroid	1.44×10-5 (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)
ENCE00000031690	1	ENCP00000003684	AMPD2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001523273 (promoter flanking region)	5	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF, H3K36me3, DNase1, H3K4me1, MEF2A, Max, PU1, H3K9ac
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF, H3K36me3, DNase1
		liver (HepG2)	PolII, Rad21, H3K4me1, H3K4me2, FOXA1, H3K27ac, CTCF, Cmyc, DNase1
		lung (IMR90)	H3K36me3, CTCF
		blood (GM12878)	DNase1, H2AZ, Rad21, CTCF
		nervous (NH-A)	CTCF, H3K4me1, H3K4me2, DNase1
		skin (NHEK)	DNase1, H3K36me3, CTCF
		NHLF	DNase1, CTCF
		Osteobl	H3K4me2, CTCF, H3K36me3
		blood (DND-41)	CTCF, H3K36me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K4me1, Max, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, Max
		A549	DNase1, H3K27ac, H3K36me3, CTCF, H3K4me2, H3K4me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
CELSR2	1	ENST00000271332 ENST00000498157	hsa-miR-4319

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CELSR2	downstream gene variant	158	ENST00000271332	NM_001408.2	ENSP00000271332	6
CELSR2	downstream gene variant	261	ENST00000489018	?	?	8
CELSR2	downstream gene variant	77	ENST00000498157	?	?	7
PSRC1	downstream gene variant	1611	ENST00000418914	?	ENSP00000413186	5
PSRC1	downstream gene variant	13	ENST00000369909	NM_001032291.2, NM_032636.7	ENSP00000358925	10
PSRC1	downstream gene variant	1442	ENST00000429031	?	ENSP00000387791	5
PSRC1	downstream gene variant	12	ENST00000369904	NM_001005290.3	ENSP00000358920	10
PSRC1	downstream gene variant	12	ENST00000369907	?	ENSP00000358923	10
PSRC1	downstream gene variant	21	ENST00000409138	?	ENSP00000474667	10
PSRC1	downstream gene variant	12	ENST00000409267	?	ENSP00000386323	10
PSRC1	downstream gene variant	1265	ENST00000474126	?	ENSP00000474682	6
PSRC1	downstream gene variant	23	ENST00000492431	?	?	10
PSRC1	downstream gene variant	2075	ENST00000471740	?	?	5
PSRC1	downstream gene variant	41	ENST00000369903	?	ENSP00000358919	9
PSRC1	downstream gene variant	1860	ENST00000459765	?	?	5

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CELSR2	ENST00000271332	NM_001408.2	ENSP00000271332	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CELSR2	ENST00000498157	?	3