Locus 28 (rs651007)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,141,870-136,155,000
block size	13,131 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.489 [-4.02 – 0.587]	**gene(s) hit or close-by**	ABO
phastCons	μ = 0.073 [0.002 – 0.373]	**eQTL gene(s)**	ABO, GBGT1, SURF1, SURF6
GERP++	μ = -0.111 [-1.2 – 0.495]	**potentially regulated gene(s)**	ADAMTSL2, CELP, DBH, RALGDS, SARDH
CADD score	μ = 4.265 [0.015 – 8.406]	**disease gene(s)**	SURF1, DBH, ADAMTSL2, SARDH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Iron status biomarkers (ferritin levels)	$<1.00\times10^{-8}$	GWAS Catalog	25352340	1
Blood metabolite levels	$<6.00\times10^{-20}$	GWAS Catalog	24816252	1
Blood metabolite ratios	$<6.00\times10^{-34}$	GWAS Catalog	24816252	2
Urinary metabolites (H-NMR features)	$<1.00\times10^{-28}$	GWAS Catalog	24586186	1
Coronary artery disease or large artery stroke	$<3.00\times10^{-8}$	GWAS Catalog	24262325	1
Coronary Artery Disease	$<2.00\times10^{-7}$	GWAS Catalog	24262325	1
Coronary artery disease or ischemic stroke	$<2.00\times10^{-9}$	GWAS Catalog	24262325	1
Serum alkaline phosphatase levels	$<1.00\times10^{-56}$	GWAS Catalog	24094242	1
End-stage coagulation	$<2.00\times10^{-25}$	GWAS Catalog	23381943	1
Red blood cell traits	$<9.00\times10^{-18}$	GWAS Catalog	23222517	1
Venous thromboembolism	$<3.00\times10^{-16}$	GWAS Catalog	22672568	1
Liver enzyme levels (alkaline phosphatase)	$<3.00\times10^{-123}$	GWAS Catalog	22001757	1
Metabolite levels	$<6.00\times10^{-9}$	GWAS Catalog	21909109	1
Coronary heart disease	$<4.00\times10^{-14}$	GWAS Catalog	21378990	1
Cholesterol, total	$<9.00\times10^{-21}$	GWAS Catalog	20686565	1
LDL cholesterol	$<8.00\times10^{-22}$	GWAS Catalog	20686565	1
Soluble levels of adhesion molecules	$<2.00\times10^{-41}$	GWAS Catalog	20167578	1
E-selectin levels	$<2.00\times10^{-82}$	GWAS Catalog	20147318	1
Red blood cell count	$<3.00\times10^{-12}$	GWAS Catalog	20139978	1
Hematological and biochemical traits	$<1.00\times10^{-11}$	GWAS Catalog	20139978	1
Angiotensin-converting enzyme activity	$<3.00\times10^{-8}$	GWAS Catalog	20066004	1
Soluble E-selectin levels	$<1.00\times10^{-29}$	GWAS Catalog	19729612	1
ADpSGEGDFXAEGGGVR*	6.34×10^{-20}	Metabolomics GWAS Server	24816252	3
E-selectin	1.33×10^{-29}	dbGaP	pha002871	2

Disease gene annotation

gene	trait	source DB	source entry/link
SURF1	LEIGH SYNDROME	OMIM	MIM:256000
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY, CONGENITAL	OMIM	MIM:223360
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1	OMIM	MIM:231050
SARDH	SARCOSINEMIA	OMIM	MIM:268900
SURF1	Complex IV deficiency	DECIPHER	MIM:220110
SURF1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
SURF1	Fatal infantile cytochrome C oxidase deficiency	OrphaNet	OrphaNet:1561
SURF1	Leigh syndrome with cardiomyopathy	OrphaNet	OrphaNet:70474
SURF1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241
SURF1	SURF1-related Charcot-Marie-Tooth disease type 4	OrphaNet	OrphaNet:391351
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY	OrphaNet	OrphaNet:230
ADAMTSL2	Geleophysic dysplasia	OrphaNet	OrphaNet:2623
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	adrenal gland	2.57×10^{-6} (p-value)	GTEx Portal V6	3
SURF1	?	ENSG00000148290	atrial appendage	4.13×10^{-6} (p-value)	GTEx Portal V6	1
ABO	?	ENSG00000175164	blood	6.42×10^{-10} (p-value)	GTEx Portal V6	7
GBGT1	ENST00000372040	ILMN_1652906	blood	1.60×10^{-6} (p-value)	Westra et al.	3
GBGT1	ENST00000470431					
GBGT1	ENST00000472281					
GBGT1	ENST00000372043					
GBGT1	ENST00000540636					
SURF6	ENST00000372022	ILMN_1778032	blood	8.04×10^{-7} (p-value)	Westra et al.	3
SURF6	ENST00000468290					
SURF6	ENST00000372022	ILMN_1778032	monocyte	4.67×10^{-5} (p-value)	Fairfax et al.	1
SURF6	ENST00000468290		b-cell	1.52×10^{-4} (p-value)	Fairfax et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000558528	1	ENCP00000060534	SARDH
		ENCP00000060486	CELP
		ENCP00000060489	RALGDS
		ENCP00000060527	ADAMTSL2
		ENCP00000060525	ADAMTSL2
		ENCP00000060535	SARDH
		ENCP00000060529	DBH

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000251886 (promoter)	2	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, Gabp, H3K4me2, H3K4me3, H3K27me3, DNase1
		HSMMtube	H3K27me3

		Osteobl	H3K27me3
		blood (K562)	ZBTB7A, H3K27ac, H3K9ac, Cmyc, H3K79me2, H3K4me3, DNase1, CTCF, H3K4me2, PolII, CTCFL, H2AZ
		skin (NHDF-AD)	H3K27me3
		breast (HMEC)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001734091 e! (TF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	PolII, H3K9ac, DNase1
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001734092 e! (enhancer)	1	HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	DNase1
		skin (NHDF-AD)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABO e!	upstream gene variant	1140	ENST00000538324 e!	?	ENSP00000483018 e!	5
ABO e!	upstream gene variant	1140	ENST00000611156 e!	NM_020469.2	ENSP00000483265 e!	5
ABO e!	upstream gene variant	1153	ENST00000453660 e!	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO e!	ENST00000538324 e!	?	ENSP00000483018 e!	3
ABO e!	ENST00000611156 e!	NM_020469.2	ENSP00000483265 e!	3
ABO e!	ENST00000453660 e!	?	?	3