Locus 69 (rs8176749)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,115,176-136,184,526
block size	69,351 bp
variant count	25 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.373 [-2.969 – 1.925]	gene(s) hit or close-by	ABO, LCN1P2, Y_RNA
phastCons	μ = 0.069 [0 – 0.636]	eQTL gene(s)	ABO, MED22, OBP2B, SLC2A6
GERP++	μ = -0.804 [-8.76 – 2.51]	potentially regulated gene(s)	ABO, ADAMTS13, ADAMTS13, ADAMTSL2, AK8, C9orf9, CELP, DBH, GFI1B, GTF3C5, OBP2B, OBP2B, RALGDS, REXO4, REXO4, SARDH, SURF1, SURF1, SURF2, SURF2, SURF4, SURF4
CADD score	μ = 3.314 [0.003 – 16.8]	disease gene(s)	SURF1, , DBH, ADAMTSL2, ADAMTS13, SARDH, GFI1B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Intraocular pressure	$<3.00\times10^{-11}$	GWAS Catalog	25173106	1
High serum lipase activity	$<1.00\times10^{-22}$	GWAS Catalog	25028398	1
Elevated serum carcinoembryonic antigen levels	$<2.00\times10^{-24}$	GWAS Catalog	24941225	1
Urinary metabolites (H-NMR features)	$<4.00\times10^{-12}$	GWAS Catalog	24586186	1
End-stage coagulation	$<2.00\times10^{-17}$	GWAS Catalog	23381943	1
Tumor biomarkers	$<7.00\times10^{-105}$	GWAS Catalog	23300138	1
Coagulation factor levels	$<2.00\times10^{-138}$	GWAS Catalog	23267103	1
Mean corpuscular hemoglobin concentration	$<4.00\times10^{-8}$	GWAS Catalog	20139978	1
Parkinson disease	8.66×10^{-6}	dbGaP	pha002868	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Abo blood group system, b(a) phenotype	association	ClinVar	RCV000019312.3	1
ABO BLOOD GROUP SYSTEM, CIS-AB PHENOTYPE	OMIM curated	OMIM	MIM:110300	1

Disease gene annotation

gene	trait	source DB	source entry/link
SURF1	Complex IV deficiency	DECIPHER	MIM:220110
SURF1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
	Complex IV deficiency	DECIPHER	MIM:220110
	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY, CONGENITAL	OMIM	MIM:223360
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1	OMIM	MIM:231050
SURF1	LEIGH SYNDROME	OMIM	MIM:256000
ADAMTS13	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL	OMIM	MIM:274150
SARDH	SARCOSINEMIA	OMIM	MIM:268900
GFI1B	BLEEDING DISORDER, PLATELET-TYPE, 17	OMIM	MIM:187900
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY	OrphaNet	OrphaNet:230

ADAMTSL2	Geleophysic dysplasia	OrphaNet	OrphaNet:2623	orphanet
SURF1	Fatal infantile cytochrome C oxidase deficiency	OrphaNet	OrphaNet:1561	orphanet
SURF1	Leigh syndrome with cardiomyopathy	OrphaNet	OrphaNet:70474	orphanet
SURF1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241	orphanet
SURF1	SURF1-related Charcot-Marie-Tooth disease type 4	OrphaNet	OrphaNet:391351	orphanet
ADAMTS13	Congenital thrombotic thrombocytopenic purpura due to ADAMTS-13 deficiency	OrphaNet	OrphaNet:93583	orphanet
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129	orphanet
GFI1B	GRAY PLATELET SYNDROME	OrphaNet	OrphaNet:721	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)
ABO	missense variant	ENST00000538324	?	ENSP00000483018	3	3			3
ABO	missense variant	ENST00000611156	NM_020469.2	ENSP00000483265	3	3			3

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	stomach	1.87×10^{-11} (p-value)	GTEx Portal V6	23
ABO	?	ENSG00000175164	lung	1.34×10^{-22} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	tibial nerve	2.83×10^{-18} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	thyroid	2.25×10^{-34} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	esophagus mucosa	4.41×10^{-26} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	skeletal muscle	2.99×10^{-11} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	transverse colon	2.25×10^{-8} (p-value)	GTEx Portal V6	22
ABO	?	ENSG00000175164	unexposed skin	1.58×10^{-26} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	sun exposed skin	4.86×10^{-27} (p-value)	GTEx Portal V6	24
OBP2B	?	ENSG00000171102	sun exposed skin	5.09×10^{-8} (p-value)	GTEx Portal V6	24
ABO	?	ENSG00000175164	subcutaneous adipocytes	7.24×10^{-10} (p-value)	GTEx Portal V6	21
ABO	?	ENSG00000175164	muscularis mucosae	1.92×10^{-7} (p-value)	GTEx Portal V6	19
ABO	?	ENSG00000175164	visceral adipocytes	2.37×10^{-8} (p-value)	GTEx Portal V6	17
ABO	?	ENSG00000175164	pancreas	7.12×10^{-7} (p-value)	GTEx Portal V6	15
ABO	?	ENSG00000175164	testis	4.43×10^{-9} (p-value)	GTEx Portal V6	20
ABO	?	ENSG00000175164	left ventricle	3.30×10^{-8} (p-value)	GTEx Portal V6	18
MED22	ENST00000614493	ILMN_1697218	monocyte	1.69×10^{-4} (p-value)	Fairfax et al.	2
MED22	ENST00000610672					
MED22	ENST00000343730					
MED22	ENST00000610888					

SLC2A6	ENST00000371899	ILMN_1778321	blood	5.84×10-7 (p-value)	Westra et al.	6
SLC2A6	ENST00000485978					
SLC2A6	ENST00000371897					
ABO	?	ENSG00000175164	adrenal gland	2.08×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000859024	1	ENST00000538324, ENST00000453660	ABO

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000558431	1	ENCP00000060477	GFI1B
		ENCP00000060525	ADAMTSL2
		ENCP00000060529	DBH
		ENCP00000060490	RALGDS
		ENCP00000060486	CELP
		ENCP00000060489	RALGDS
		ENCP00000060518	ADAMTS13 ADAMTS13
		ENCP00000060527	ADAMTSL2
		ENCP00000060535	SARDH
		ENCP00000060507	SURF2 SURF2 SURF1 SURF1
		ENCP00000060492	RALGDS
ENCE00000558621	1	ENCP00000060495	OBP2B OBP2B
ENCE00000558373	3	ENCP00000060477	GFI1B
		ENCP00000060471	AK8 C9orf9
		ENCP00000060525	ADAMTSL2
		ENCP00000060529	DBH
		ENCP00000060516	REXO4 REXO4
		ENCP00000060509	SURF4 SURF4
		ENCP00000060490	RALGDS
		ENCP00000060486	CELP
		ENCP00000060489	RALGDS
		ENCP00000060518	ADAMTS13 ADAMTS13
		ENCP00000060527	ADAMTSL2
		ENCP00000060535	SARDH
		ENCP00000060507	SURF2 SURF2 SURF1 SURF1
		ENCP00000060492	RALGDS
ENCE00000558506	1	ENCP00000060483	GTF3C5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001734089 (enhancer)	2	liver (HepG2)	H3K4me1, H3K4me2
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K4me3, H3K4me2
		blood (K562)	H3K4me2, H2AZ
		skin (NHDF-AD)	H3K4me2
ENSR00000452813 (promoter flanking region)	2	cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, PolII, DNase1
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
ENSR00001475637 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, Brg1, Ini1, PolII
		A549	H3K27me3
ENSR00001475641 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABO	downstream gene variant	2696	ENST00000453660	?	?	1
ABO	downstream gene variant	248	ENST00000538324	?	ENSP00000483018	7
ABO	downstream gene variant	848	ENST00000611156	NM_020469.2	ENSP00000483265	6
LCN1P2	upstream gene variant	103	ENST00000372030	?	?	1
Y_RNA	downstream gene variant	4516	ENST00000364973	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	2	2	2
ABO	ENST00000538324	?	ENSP00000483018	2	2	2

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000453660	?	?	7
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	7
ABO	ENST00000538324	?	ENSP00000483018	7

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ABO	ENST00000453660	?	12