Locus 442 (rs7857390)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,128,546-136,147,295
block size	18,750 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.281 [-2.374 – 0.721]	gene(s) hit or close-by	ABO
phastCons	μ = 0.112 [0 – 0.656]	eQTL gene(s)	ABO, GBGT1, SLC2A6, SURF1
GERP++	μ = 0.030 [-2.56 – 1.11]	potentially regulated gene(s)	–
CADD score	μ = 2.951 [0.029 – 13.47]	disease gene(s)	SURF1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
ADSGEGDFXAEGGGVR*	1.17×10-6	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link
SURF1	LEIGH SYNDROME	OMIM	MIM:256000
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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SURF1	?	ENSG00000148290	sun exposed skin	3.29×10-6 (p-value)	GTEx Portal V6	12
ABO	?	ENSG00000175164	esophagus mucosa	2.75×10-7 (p-value)	GTEx Portal V6	12
ABO	?	ENSG00000175164	skeletal muscle	4.13×10-6 (p-value)	GTEx Portal V6	7
GBGT1	ENST00000372040	ILMN_1652906	blood	2.77×10-6 (p-value)	Westra et al.	2
GBGT1	ENST00000470431					
GBGT1	ENST00000472281					
GBGT1	ENST00000372043					
GBGT1	ENST00000540636					
SLC2A6	ENST00000371899	ILMN_1778321	blood	7.27×10-5 (p-value)	Westra et al.	2
SLC2A6	ENST00000485978					
SLC2A6	ENST00000371897					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
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

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABO	downstream gene variant	2443	ENST00000538324	?	ENSP00000483018	1
ABO	downstream gene variant	2017	ENST00000611156	NM_020469.2	ENSP00000483265	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000453660	?	?	12
ABO	ENST00000538324	?	ENSP00000483018	12
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	12

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ABO	ENST00000453660	?	1