Locus 269 (rs630510)

SNiPAcard

Block annotations

Block info

genomic range	chr9:136,149,581-136,150,484
block size	904 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.148 [-1.212 – 0.562]	**gene(s) hit or close-by**	ABO
phastCons	μ = 0.009 [0.001 – 0.016]	**eQTL gene(s)**	ABO, MED22
GERP++	μ = -0.746 [-1.19 – 0.225]	**potentially regulated gene(s)**	ADAMTSL2, CELP, DBH, RALGDS, SARDH
CADD score	μ = 6.844 [5.434 – 8.361]	**disease gene(s)**	DBH, ADAMTSL2, SARDH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
ADpSGEGDFXAEGGGVR*	3.53×10^{-9}	Metabolomics GWAS Server	24816252	1
Pancreatic neoplasms	4.70×10^{-6}	dbGaP	pha002889	1
Pancreatic neoplasms	4.28×10^{-5}	dbGaP	pha002874	1
E-selectin	9.65×10^{-13}	dbGaP	pha002871	1

Disease gene annotation

gene	trait	source DB	source entry/link
DBH	DOPAMINE BETA-HYDROXYLASE DEFICIENCY, CONGENITAL	OMIM	MIM:223360
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1	OMIM	MIM:231050
SARDH	SARCOSINEMIA	OMIM	MIM:268900
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	pancreas	9.62×10^{-10} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	adrenal gland	6.23×10^{-12} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	tibial nerve	9.48×10^{-9} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	transverse colon	1.58×10^{-7} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	esophagus mucosa	5.75×10^{-11} (p-value)	GTEx Portal V6	4
ABO	?	ENSG00000175164	thyroid	6.65×10^{-13} (p-value)	GTEx Portal V6	4
MED22	ENST00000614493	ILMN_1697218	monocyte	1.74×10^{-5} (p-value)	Fairfax et al.	1
MED22	ENST00000610672		b-cell	1.18×10^{-5} (p-value)	Fairfax et al.	1
MED22	ENST00000343730					

MED22	ENST00000610888

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)	4	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

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000538324	?	ENSP00000483018	4
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	4
ABO	ENST00000453660	?	?	4