SNiPAcard

Block annotations

Block info

genomic range	chr9:136,137,065-136,149,500
block size	12,436 bp
variant count	35 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.587 [-2.293 – 0.687]	**gene(s) hit or close-by**	ABO
phastCons	μ = 0.133 [0 – 0.646]	**eQTL gene(s)**	ABO, GBGT1
GERP++	μ = -0.006 [-0.989 – 1.37]	**potentially regulated gene(s)**	ABO, ABO, AK8, SARDH, TSC1, TSC1
CADD score	μ = 2.446 [0.195 – 8.142]	**disease gene(s)**	TSC1, SARDH

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
ADpSGEGDFXAEGGGVR*	9.66×10^{-14}	Metabolomics GWAS Server	24816252	11

Disease gene annotation

gene	trait	source DB	source entry/link
TSC1	FOCAL CORTICAL DYSPLASIA OF TAYLOR	OMIM	MIM:607341
TSC1	TUBEROUS SCLEROSIS 1	OMIM	MIM:191100
SARDH	SARCOSINEMIA	OMIM	MIM:268900
TSC1	tuberous sclerosis type 1 (TSC1)	DECIPHER	MIM:191100
TSC1	Tuberous sclerosis	OrphaNet	OrphaNet:805
TSC1	LYMPHANGIOLEIOMYOMATOSIS	OrphaNet	OrphaNet:538
TSC1	Isolated focal cortical dysplasia type IIb	OrphaNet	OrphaNet:269008
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	pancreas	1.36×10^{-13} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	lung	7.77×10^{-12} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	atrial appendage	8.21×10^{-9} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	thyroid	8.24×10^{-16} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	skeletal muscle	5.22×10^{-8} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	transverse colon	1.80×10^{-9} (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	sun exposed skin	4.00×10^{-6} (p-value)	GTEx Portal V6	26
ABO	?	ENSG00000175164	unexposed skin	5.93×10^{-6} (p-value)	GTEx Portal V6	1
ABO	?	ENSG00000175164	ovary	4.63×10^{-13} (p-value)	GTEx Portal V6	34

ABO	?	ENSG00000175164	visceral adipocytes	2.24×10-11 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	tibial nerve	1.62×10-9 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	vagina	2.69×10-9 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	esophagus mucosa	1.79×10-32 (p-value)	GTEx Portal V6	34
GBGT1 GBGT1 GBGT1 GBGT1 GBGT1	ENST00000372040 ENST00000470431 ENST00000472281 ENST00000372043 ENST00000540636	ILMN_1652906	blood	5.72×10-7 (p-value)	Westra et al.	11
ABO	?	ENSG00000175164	adrenal gland	2.39×10-21 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	testis	4.95×10-9 (p-value)	GTEx Portal V6	34
ABO	?	ENSG00000175164	subcutaneous adipocytes	2.56×10-6 (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)
ENCE00000558384	1	ENCP00000060472	TSC1 TSC1
ENCE00000558361	1	ENCP00000060497	ABO ABO
		ENCP00000060532	SARDH
		ENCP00000060469	AK8

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000251881 (TF binding site)	1	lung (IMR90)	H3K27me3
		blood (K562)	H3K36me3, DNase1, Brg1, Ini1
		A549	H3K27me3
ENSR00001475637 (TF binding site)	5	embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K36me3, H3K4me2, H3K9ac, H3K27ac, DNase1, Brg1, Ini1, PolII
		A549	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000538324	?	ENSP00000483018	35
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	35
ABO	ENST00000453660	?	?	35