SNiPAcard

Block annotations

Block info

genomic range	chr9:136,128,467-136,132,873
block size	4,407 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.049 [-2.334 – 0.014]	**gene(s) hit or close-by**	ABO
phastCons	μ = 0.039 [0 – 0.19]	**eQTL gene(s)**	ABO, SURF6
GERP++	μ = -1.687 [-8.39 – 0.522]	**potentially regulated gene(s)**	–
CADD score	μ = 2.053 [0.112 – 6.094]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Serum alkaline phosphatase levels	$<3.00\times10^{-21}$	GWAS Catalog	24094242	1
ADpSGEGDFXAEGGGVR*	2.01×10^{-7}	Metabolomics GWAS Server	24816252	3
E-selectin	7.24×10^{-7}	dbGaP	pha002871	2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABO	?	ENSG00000175164	blood	6.00×10^{-6} (p-value)	GTEx Portal V6	3
SURF6	ENST00000372022	ILMN_1778032	skin	1.54×10^{-5} (p-value)	MuTHER consortium	3
SURF6	ENST00000468290					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABO	downstream gene variant	96	ENST00000538324	?	ENSP00000483018	5
ABO	downstream gene variant	551	ENST00000611156	NM_020469.2	ENSP00000483265	4

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	T	acG/acA	1
ABO	ENST00000538324	?	ENSP00000483018	T	acG/acA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABO	ENST00000453660	?	?	5
ABO	ENST00000611156	NM_020469.2	ENSP00000483265	5
ABO	ENST00000538324	?	ENSP00000483018	5

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	?	6