Locus 377 (rs983545)

SNiPAcard

Block annotations

Block info

genomic range	chr3:16,947,451-16,972,211
block size	24,761 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.151 [-1.745 – 1.014]	gene(s) hit or close-by	MIR3714, PLCL2
phastCons	μ = 0.033 [0 – 0.422]	eQTL gene(s)	PLCL2
GERP++	μ = -1.075 [-7.38 – 3.22]	potentially regulated gene(s)	PLCL2
CADD score	μ = 3.650 [0.087 – 10.54]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Primary biliary cirrhosis	<2.00×10-8	GWAS Catalog	21399635	1
caproate (6:0)	8.64×10-6	Metabolomics GWAS Server	24816252	2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PLCL2	ENST00000432376	ILMN_1737025	adipocyte	3.87×10-8 (p-value)	MuTHER consortium	12
PLCL2	ENST00000615277					
PLCL2	?	ENSG00000154822	transformed fibroblasts	4.04×10-7 (p-value)	GTEx Portal V6	16
PLCL2	?	ENSG00000154822	blood	2.38×10-10 (p-value)	GTEx Portal V6	17
PLCL2	ENST00000615277	ILMN_1737025	monocyte	4.25×10-9 (p-value)	Fairfax et al.	2
PLCL2	ENST00000432376		b-cell	2.47×10-4 (p-value)	Fairfax et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000331323	1	ENCP00000035615	PLCL2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001673018 (enhancer)	2	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		blood (GM12878)	H3K79me2
		blood (K562)	H3K27me3
ENSR00001673019 (promoter flanking region)	6	monocytes (Monocytes-CD14+)	H4K20me1
		endothelium (HUVEC)	PolII
		blood (GM12878)	H3K79me2

		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00001477059 e! (CTCF binding site)	4	monocytes (Monocytes-CD14+)	H4K20me1
		Osteobl	H3K4me2
		blood (GM12878)	H3K79me2
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MIR3714 e!	upstream gene variant	2477	ENST00000579507 e!	?	?	1
PLCL2 e!	upstream gene variant	2371	ENST00000432376 e!	NM_015184.5	ENSP00000412836 e!	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PLCL2 e!	ENST00000460467 e!	?	?	18
PLCL2 e!	ENST00000615277 e!	NM_001144382.1	ENSP00000478458 e!	18