SNiPAcard

Block annotations

Block info

genomic range	chr10:54,531,685-54,536,083
block size	4,399 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.723 [-2.66 – 0.688]	**gene(s) hit or close-by**	MBL2
phastCons	μ = 0.001 [0 – 0.006]	**eQTL gene(s)**	MBL2
GERP++	μ = -0.672 [-4.14 – 2.24]	**potentially regulated gene(s)**	DKK1, MBL2, MBL2
CADD score	μ = 2.325 [0.134 – 5.949]	**disease gene(s)**	MBL2, DKK1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1-arachidonoylglycerophosphocholine*	7.59×10^{-5}	Metabolomics GWAS Server	24816252	2

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CR952185	1

Disease gene annotation

gene	trait	source DB	source entry/link
MBL2	MANNOSE-BINDING PROTEIN DEFICIENCY	OMIM	MIM:614372
MBL2	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
DKK1	Idiopathic juvenile osteoporosis	OrphaNet	OrphaNet:85193

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MBL2	?	ENSG00000165471	tibial nerve	9.48×10^{-9} (p-value)	GTEx Portal V6	6
MBL2	?	ENSG00000165471	liver	3.29×10^{-10} (p-value)	GTEx Portal V6	6
MBL2	ENST00000373968	ILMN_1762464	liver	$<2.2\times10^{-16}$ (p-value)	Innocenti et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000074894	1	ENCP00000008487	DKK1
		ENCP00000008488	DKK1
		ENCP00000008495	MBL2 MBL2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MBL2	upstream gene variant	225	ENST00000373968	NM_000242.2	ENSP00000363079	6