Locus 257 (rs2506)

SNiPAcard

Block annotations

Block info

genomic range	chr10:54,504,733-54,526,622
block size	21,890 bp
variant count	57 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.534 [-3.733 – 1.202]	gene(s) hit or close-by	MBL2, RP11-556E13.1
phastCons	μ = 0.013 [0 – 0.269]	eQTL gene(s)	PRKRIRP3
GERP++	μ = -0.496 [-5.36 – 1.66]	potentially regulated gene(s)	RP11-556E13.1
CADD score	μ = 2.669 [0.013 – 9.19]	disease gene(s)	MBL2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
theophylline	3.37×10^{-5}	Metabolomics GWAS Server	24816252	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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PRKRIRP3	?	ENSG00000227972	tibial nerve	2.66×10^{-7} (p-value)	GTEx Portal V6	39

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000074939	1	ENCP00000008494	RP11-556E13.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000355146 (CTCF binding site)	5	HSMMtube	H3K27me3
		liver (HepG2)	CTCF
ENSR00001596434 (promoter flanking region)	1	liver (HepG2)	H3K4me2, H3K9ac, H3K27ac, H3K4me1
		HSMMtube	H3K27me3
ENSR00001016746 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	FOSL2, Jund, H3K4me1, p300, H3K4me2, H3K9ac, H3K27ac
		HSMMtube	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	DNase1

		muscle (HSMM)	DNase1
ENSR00001596435 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	FOSL2, Jund, H3K4me1, p300, H3K4me2, H3K9ac, H3K27ac
		A549	H3K27me3
		nervous (NH-A)	DNase1
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MBL2	downstream gene variant	482	ENST00000373968	NM_000242.2	ENSP00000363079	9
RP11-556E13.1	upstream gene variant	421	ENST00000443523	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RP11-556E13.1	ENST00000443523	?	?	36

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MBL2	ENST00000373968	NM_000242.2	ENSP00000363079	8