Locus 93 (rs7588285)

SNiPAcard

Block annotations

Block info

genomic range	chr2:3,639,909-3,648,186
block size	8,278 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.830 [-1.637 – 0.331]	gene(s) hit or close-by	AC010907.2, COLEC11
phastCons	μ = 0.022 [0 – 0.08]	eQTL gene(s)	ALLC, COLEC11, RNASEH1-AS1
GERP++	μ = 0.320 [-0.397 – 2.32]	potentially regulated gene(s)	–
CADD score	μ = 4.127 [0.417 – 5.959]	disease gene(s)	COLEC11

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
COLEC11	3MC SYNDROME 2	OMIM	MIM:265050
COLEC11	3MC SYNDROME 2	DECIPHER	MIM:265050
COLEC11	Craniofacial-ulnar-renal syndrome	OrphaNet	OrphaNet:293843

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ALLC	?	ENSG00000151360	liver	1.92×10^{-6} (p-value)	GTEx Portal V6	2
RNASEH1-AS1	?	ENSG00000234171	thyroid	2.37×10^{-6} (p-value)	GTEx Portal V6	3
COLEC11	?	ENSG00000118004	pancreas	2.78×10^{-7} (p-value)	GTEx Portal V6	3
RNASEH1-AS1	?	ENSG00000234171	tibial nerve	9.49×10^{-6} (p-value)	GTEx Portal V6	1
COLEC11	ENST00000382062	ILMN_1735192	liver	1.68×10^{-11} (p-value)	Innocenti et al.	1
COLEC11	ENST00000402922					
COLEC11	ENST00000403096					
COLEC11	ENST00000402794					
COLEC11	ENST00000487365					
COLEC11	ENST00000416132					
COLEC11	ENST00000460971					
COLEC11	ENST00000349077					
COLEC11	ENST00000404205					
COLEC11	ENST00000236693					
COLEC11	ENST00000418971					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors

ENSR00000584312 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, TAF1, Yy1, Rad21, CTCF, H3K27me3
		endothelium (HUVEC)	DNase1
		Osteobl	H3K27me3, H3K4me2
		blood (GM12878)	H3K36me3
		blood (K562)	PolII, Max, Rad21, CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC010907.2	downstream gene variant	2801	ENST00000456450	?	?	2
COLEC11	upstream gene variant	383	ENST00000416132	?	ENSP00000410919	5
COLEC11	upstream gene variant	321	ENST00000349077	NM_001255982.1, NM_024027.4	ENSP00000339168	5
COLEC11	upstream gene variant	184	ENST00000460971	?	?	5
COLEC11	upstream gene variant	61	ENST00000382062	NM_001255983.1, NM_001255984.1	ENSP00000371494	5
COLEC11	upstream gene variant	1310	ENST00000418971	NM_001255985.1	ENSP00000411770	2
COLEC11	upstream gene variant	278	ENST00000236693	NM_199235.2	ENSP00000236693	5
COLEC11	upstream gene variant	184	ENST00000487365	?	?	5
COLEC11	upstream gene variant	65	ENST00000419002	?	?	5
COLEC11	upstream gene variant	1310	ENST00000438814	?	ENSP00000393167	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
COLEC11	ENST00000416132	?	ENSP00000410919	2
COLEC11	ENST00000460971	?	?	2
COLEC11	ENST00000349077	NM_001255982.1, NM_024027.4	ENSP00000339168	2
COLEC11	ENST00000487365	?	?	2
COLEC11	ENST00000419002	?	?	2
COLEC11	ENST00000382062	NM_001255983.1, NM_001255984.1	ENSP00000371494	2
COLEC11	ENST00000236693	NM_199235.2	ENSP00000236693	2