Locus 299 (rs6547631)

SNiPAcard

Block annotations

Block info

genomic range	chr2:85,909,026-85,912,870
block size	3,845 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.129 [-0.867 – 0.579]	gene(s) hit or close-by	AC012454.4, GNLY
phastCons	μ = 0.022 [0.001 – 0.086]	eQTL gene(s)	AC012454.4, ATOH8, GNLY
GERP++	μ = -0.212 [-0.761 – 0.388]	potentially regulated gene(s)	–
CADD score	μ = 4.218 [1.561 – 8.197]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC012454.4	?	ENSG00000203363	blood	5.76×10-6 (p-value)	GTEx Portal V6	1
GNLY	?	ENSG00000115523	blood	9.30×10-12 (p-value)	GTEx Portal V6	3
GNLY	?	ENSG00000115523	left ventricle	3.43×10-7 (p-value)	GTEx Portal V6	3
GNLY	?	ENSG00000115523	subcutaneous adipocytes	4.26×10-7 (p-value)	GTEx Portal V6	3
?	?	ILMN_1790692	monocyte	5.43×10-20 (p-value)	Zeller et al.	1
ATOH8	ENST00000469442	ILMN_1654065	monocyte	1.82×10-75 (p-value)	Zeller et al.	1
ATOH8	ENST00000306279					
ATOH8	ENST00000489682					
GNLY	ENST00000489214	ILMN_1708779	monocyte	5.56×10-12 (p-value)	Zeller et al.	1
GNLY	ENST00000409696					
GNLY	ENST00000534351					
GNLY	ENST00000489980					
GNLY	ENST00000533041					
GNLY	ENST00000482900					
GNLY	ENST00000470974					
GNLY	ENST00000464298					
GNLY	ENST00000263863					
GNLY	ENST00000524600					
GNLY	?	ENSG00000115523	thyroid	1.18×10-6 (p-value)	GTEx Portal V6	2
GNLY	?	ENSG00000115523	lung	6.10×10-6 (p-value)	GTEx Portal V6	1
GNLY	?	ENSG00000115523	sun exposed skin	8.77×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000593767	1	cervix (HeLa-S3)	H3K4me2, H3K27ac, DNase1

(enhancer)	embryonic stem cell (H1ESC)	H3K27me3
	HSMMtube	DNase1
	liver (HepG2)	H3K27me3
	blood (K562)	H3K27me3
	blood (DND-41)	H3K27me3
	muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC012454.4 e!	downstream gene variant	306	ENST00000443948 e!	?	?	4
GNLY e!	upstream gene variant	669	ENST00000488945 e!	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GNLY e!	ENST00000488945 e!	?	?	2