Locus 290 (rs6556937)

SNiPAcard

Block annotations

Block info

genomic range	chr5:95,985,282-95,985,282
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.268	gene(s) hit or close-by	CAST
phastCons	0.253	eQTL gene(s)	CAST, CTC-506B8.1, ERAP1
GERP++	2.41	potentially regulated gene(s)	–
CADD score	8.833	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ERAP1	?	ENSG00000164307	pancreas	5.79×10^{-6} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	lung	4.26×10^{-10} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	lung	8.60×10^{-18} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	transformed fibroblasts	7.47×10^{-10} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	transformed fibroblasts	8.00×10^{-15} (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	blood	3.77×10^{-7} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	blood	2.55×10^{-13} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	blood	1.27×10^{-20} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	breast	6.66×10^{-7} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	tibial artery	4.40×10^{-14} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	tibial artery	1.14×10^{-13} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	thyroid	2.03×10^{-7} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	thyroid	7.36×10^{-11} (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	skeletal muscle	3.16×10^{-6} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	EBV lymphocytes	1.74×10^{-6} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	EBV lymphocytes	2.48×10^{-6} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	ovary	2.54×10^{-8} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	ovary	1.78×10^{-7} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	aorta	1.54×10^{-6} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	aorta	7.30×10^{-9} (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	subcutaneous adipocytes	2.85×10^{-10} (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	subcutaneous adipocytes	5.43×10^{-12} (p-value)	GTEx Portal V6	1

?	?	ILMN_1717234	monocyte	1.20×10-13 (p-value)	Zeller et al.	1
CTC-506B8.1	?	ENSG00000249180	visceral adipocytes	1.16×10-8 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	visceral adipocytes	2.65×10-12 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	sigmoid colon	1.58×10-7 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	tibial nerve	4.62×10-10 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	tibial nerve	2.36×10-20 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	coronary artery	8.08×10-8 (p-value)	GTEx Portal V6	1
CTC-506B8.1	?	ENSG00000249180	coronary artery	4.82×10-7 (p-value)	GTEx Portal V6	1
ERAP1	?	ENSG00000164307	esophagus mucosa	1.48×10-5 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	adrenal gland	9.17×10-7 (p-value)	GTEx Portal V6	1
CAST	?	ENSG00000153113	cerebellum	2.42×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001285945 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1, DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CAST	ENST00000505143	?	ENSP00000422957	1