Locus 32 (rs3827278)

SNiPAcard

Block annotations

Block info

genomic range	chr22:17,595,915-17,596,322
block size	408 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.080 [-0.258 – 0.328]	gene(s) hit or close-by	CECR6, IL17RA
phastCons	μ = 0.000 [0 – 0.001]	eQTL gene(s)	AC006946.15, IL17RA
GERP++	μ = -0.095 [-2.8 – 1.55]	potentially regulated gene(s)	–
CADD score	μ = 2.715 [1.84 – 3.715]	disease gene(s)	IL17RA

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
IL17RA	CANDIDIASIS, FAMILIAL, 5	OMIM	MIM:613953
IL17RA	Chronic mucocutaneous candidosis	OrphaNet	OrphaNet:1334

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL17RA	?	ENSG00000177663	pancreas	6.93×10^{-9} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	muscularis mucosae	3.89×10^{-7} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	lung	1.96×10^{-7} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	atrial appendage	1.59×10^{-6} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	transformed fibroblasts	3.23×10^{-15} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	tibial artery	5.99×10^{-9} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	thyroid	3.81×10^{-11} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	skeletal muscle	7.33×10^{-10} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	unexposed skin	5.37×10^{-7} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	sun exposed skin	1.86×10^{-13} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	aorta	1.11×10^{-10} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	left ventricle	1.59×10^{-8} (p-value)	GTEx Portal V6	3
AC006946.15	ENST00000441544	ILMN_1888355	monocyte	8.94×10^{-23} (p-value)	Zeller et al.	1
IL17RA	?	ENSG00000177663	subcutaneous adipocytes	1.47×10^{-15} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	stomach	1.24×10^{-6} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	tibial nerve	4.92×10^{-9} (p-value)	GTEx Portal V6	3

IL17RA	?	ENSG00000177663	esophagus mucosa	1.03×10^{-8} (p-value)	GTEx Portal V6	3
IL17RA	?	ENSG00000177663	gastroesophageal junction	3.12×10^{-6} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CECR6	downstream gene variant	868	ENST00000331437	NM_031890.3	ENSP00000329318	3
CECR6	downstream gene variant	867	ENST00000399875	NM_001163079.1	ENSP00000382764	3

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL17RA	ENST00000612619	NM_001289905.1	ENSP00000479970	3
IL17RA	ENST00000319363	NM_014339.6	ENSP00000320936	3