

Block annotations

Block info


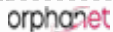
genomic range	chr22:17,595,915-17,596,322 <i>e!</i>
block size	408 bp
variant count	3 variants

Basic features

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


Trait annotations

Disease gene annotation

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

Direct effect on regulation

cis-eQTL

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

IL17RA <i>e!</i>	?	ENSG00000177663 <i>e!</i>	esophagus mucosa	1.03×10 ⁻⁸ (p-value)	GTEx Portal V6 <i>e!</i>	3
IL17RA <i>e!</i>	?	ENSG00000177663 <i>e!</i>	gastroesophageal junction	3.12×10 ⁻⁶ (p-value)	GTEx Portal V6 <i>e!</i>	3

Putative effect on regulation

Variation proximal to gene

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

Putative effect on transcript

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL17RA <i>e!</i>	ENST00000612619 <i>e!</i>	NM_001289905.1	ENSP00000479970 <i>e!</i>	3
IL17RA <i>e!</i>	ENST00000319363 <i>e!</i>	NM_014339.6	ENSP00000320936 <i>e!</i>	3

