SNiPAcard

Block annotations

Block info

genomic range	chr12:69,729,434-69,747,834
block size	18,401 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.116 [-1.544 – 0.956]	**gene(s) hit or close-by**	LYZ, RP11-1143G9.4, RP11-1143G9.5
phastCons	μ = 0.171 [0 – 0.922]	**eQTL gene(s)**	CPM, LYZ, RP11-1143G9.4, YEATS4
GERP++	μ = -1.254 [-4.79 – 3.4]	**potentially regulated gene(s)**	–
CADD score	μ = 4.343 [0.265 – 18.4]	**disease gene(s)**	LYZ

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LYZ	AMYLOIDOSIS, FAMILIAL VISCERAL	OMIM	MIM:105200
LYZ	Familial renal amyloidosis due to lysozyme variant	OrphaNet	OrphaNet:93561

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
YEATS4	ENST00000247843	ILMN_1801387	blood	8.39×10^{-6} (p-value)	MuTHER consortium	5
YEATS4	ENST00000548020		adipocyte	1.48×10^{-9} (p-value)	MuTHER consortium	5
			skin	6.04×10^{-6} (p-value)	MuTHER consortium	5
YEATS4	?	ENSG00000127337	uterus	6.55×10^{-8} (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	muscularis mucosae	9.16×10^{-13} (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	muscularis mucosae	5.32×10^{-15} (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	muscularis mucosae	4.85×10^{-10} (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	lung	4.55×10^{-34} (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	lung	3.98×10^{-11} (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	lung	2.35×10^{-33} (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	atrial appendage	7.74×10^{-13} (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	atrial appendage	1.93×10^{-8} (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	atrial appendage	2.72×10^{-10} (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	tibial artery	4.52×10^{-25} (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	tibial artery	3.94×10^{-17} (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	tibial artery	1.47×10^{-23} (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	breast	8.36×10^{-11} (p-value)	GTEx Portal V6	8

YEATS4	?	ENSG00000127337	breast	3.55×10-15 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	breast	2.27×10-10 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	blood	1.00×10-56 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	blood	1.24×10-15 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	blood	8.25×10-53 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	thyroid	1.81×10-16 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	thyroid	3.11×10-34 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	thyroid	3.44×10-15 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	skeletal muscle	3.51×10-13 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	skeletal muscle	2.36×10-31 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	skeletal muscle	3.17×10-10 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	transverse colon	5.76×10-11 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	transverse colon	5.70×10-9 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	transverse colon	1.73×10-9 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	EBV lymphocytes	1.19×10-12 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	unexposed skin	3.15×10-9 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	unexposed skin	1.21×10-19 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	unexposed skin	5.30×10-7 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	sun exposed skin	3.83×10-16 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	sun exposed skin	1.00×10-29 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	sun exposed skin	1.53×10-12 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	left ventricle	4.12×10-13 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	left ventricle	1.17×10-12 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	left ventricle	2.02×10-13 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	aorta	3.92×10-15 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	aorta	5.92×10-9 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	aorta	2.28×10-13 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	subcutaneous adipocytes	3.53×10-24 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	subcutaneous adipocytes	1.16×10-31 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	subcutaneous adipocytes	1.83×10-21 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	visceral adipocytes	2.86×10-15 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	visceral adipocytes	2.40×10-14 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	visceral adipocytes	1.56×10-13 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	stomach	3.45×10-7 (p-value)	GTEx Portal V6	4
YEATS4	?	ENSG00000127337	stomach	1.23×10-6 (p-value)	GTEx Portal V6	4
RP11-1143G9.4	?	ENSG00000257764	stomach	8.13×10-7 (p-value)	GTEx Portal V6	1

YEATS4	?	ENSG00000127337	nucleus accumbens	4.71×10-9 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	spleen	2.72×10-8 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	spleen	2.92×10-8 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	tibial nerve	5.77×10-21 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	tibial nerve	4.19×10-15 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	tibial nerve	4.78×10-18 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	coronary artery	2.17×10-6 (p-value)	GTEx Portal V6	7
YEATS4	?	ENSG00000127337	sigmoid colon	3.81×10-9 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	vagina	4.28×10-9 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	esophagus mucosa	3.72×10-8 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	esophagus mucosa	1.35×10-9 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	esophagus mucosa	4.97×10-7 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	adrenal gland	2.06×10-10 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	adrenal gland	1.12×10-8 (p-value)	GTEx Portal V6	8
RP11-1143G9.4	?	ENSG00000257764	adrenal gland	5.23×10-9 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	testis	6.72×10-7 (p-value)	GTEx Portal V6	8
YEATS4	?	ENSG00000127337	testis	1.82×10-12 (p-value)	GTEx Portal V6	4
RP11-1143G9.4	?	ENSG00000257764	testis	5.10×10-6 (p-value)	GTEx Portal V6	1
YEATS4	?	ENSG00000127337	cerebellum	4.52×10-6 (p-value)	GTEx Portal V6	8
LYZ	?	ENSG00000090382	pancreas	6.60×10-6 (p-value)	GTEx Portal V6	1
LYZ	?	ENSG00000090382	prostate	2.08×10-6 (p-value)	GTEx Portal V6	1
YEATS4	?	ENSG00000127337	caudate basal ganglia	5.11×10-7 (p-value)	GTEx Portal V6	1
YEATS4	ENST00000247843	ILMN_1801387	monocyte	1.26×10-13 (p-value)	Fairfax et al.	3
YEATS4	ENST00000548020		b-cell	1.55×10-7 (p-value)	Fairfax et al.	3
LYZ	ENST00000261267	ILMN_2162972	monocyte	8.90×10-78 (p-value)	Fairfax et al.	3
CPM	ENST00000338356	ILMN_2299795	monocyte	1.58×10-8 (p-value)	Fairfax et al.	3
CPM	ENST00000551568					
?	?	ILMN_1815205	monocyte	2.31×10-17 (p-value)	Fairfax et al.	3
			b-cell	4.27×10-18 (p-value)		
YEATS4	?	ENSG00000127337	cortex	1.95×10-6 (p-value)	GTEx Portal V6	4
YEATS4	?	ENSG00000127337	cerebellar hemisphere	3.87×10-6 (p-value)	GTEx Portal V6	4

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001464668 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
ENSR00001612777	1	NHLF	H3K27me3

(enhancer)		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1
		lung (IMR90)	H3K27me3
		A549	H3K27me3
ENSR00001612779 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1, H3K4me2, H3K27ac, H3K4me3
		liver (HepG2)	H3K27ac, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LYZ	upstream gene variant, downstream gene variant	1297	ENST00000548839	?	ENSP00000449969	4
LYZ	upstream gene variant, downstream gene variant	559	ENST00000549690	?	ENSP00000449898	3
LYZ	upstream gene variant	1252	ENST00000261267	NM_000239.2	ENSP00000261267	2
RP11-1143G9.4	downstream gene variant	602	ENST00000548900	?	?	1
RP11-1143G9.5	upstream gene variant	3772	ENST00000613291	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LYZ	ENST00000549690	?	ENSP00000449898	1
LYZ	ENST00000261267	NM_000239.2	ENSP00000261267	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LYZ	ENST00000261267	NM_000239.2	ENSP00000261267	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RP11-1143G9.4	ENST00000548900	?	1