Locus 313 (rs6442112)

SNiPAcard

Block annotations

Block info

genomic range	chr3:48,221,109-48,316,034
block size	94,926 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.964 [-2.748 – -1.105]	gene(s) hit or close-by	CAMP, CDC25A, NDUFB1P1, RNU7-128P, ZNF589
phastCons	μ = 0.002 [0 – 0.007]	eQTL gene(s)	AMT, ARIH2OS, CDC25A, FCF1P2, KLHDC8B, MRPS18AP1, NCKIPSD, NICN1, NME6, QRICH1, SCAP, TMA7, WDR6, ZNF589
GERP++	μ = -0.371 [-1.44 – 0.624]	potentially regulated gene(s)	FBXW12, MAP4, SHISA5, UQCRC1
CADD score	μ = 1.210 [0.443 – 1.735]	disease gene(s)	AMT, KLHDC8B

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899	OMIM
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899	OMIM
KLHDC8B	LYMPHOMA, HODGKIN, CLASSIC	OMIM	MIM:236000	OMIM
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860	orphanet
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857	orphanet
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NME6	?	ENSG00000172113	muscularis mucosae	3.38×10-7 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	muscularis mucosae	8.74×10-8 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	muscularis mucosae	5.58×10-6 (p-value)	GTEx Portal V6	3
NCKIPSD	?	ENSG00000213672	muscularis mucosae	5.12×10-6 (p-value)	GTEx Portal V6	2
QRICH1	?	ENSG00000198218	lung	2.11×10-6 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	lung	3.98×10-7 (p-value)	GTEx Portal V6	3
SCAP	?	ENSG00000114650	atrial appendage	4.01×10-6 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	atrial appendage	1.18×10-5 (p-value)	GTEx Portal V6	2
NME6	?	ENSG00000172113	transformed fibroblasts	1.48×10-7 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	transformed fibroblasts	4.53×10-10 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	transformed fibroblasts	3.74×10-7 (p-value)	GTEx Portal V6	2
NCKIPSD	?	ENSG00000213672	transformed fibroblasts	4.27×10-7 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	tibial artery	4.55×10-9 (p-value)	GTEx Portal V6	3

ZNF589	?	ENSG00000164048	tibial artery	4.23×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	tibial artery	1.31×10-5 (p-value)	GTEx Portal V6	1
CDC25A	?	ENSG00000164045	tibial artery	1.43×10-5 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	blood	2.78×10-8 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	blood	2.49×10-6 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	thyroid	3.43×10-8 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	thyroid	1.66×10-5 (p-value)	GTEx Portal V6	2
NCKIPSD	?	ENSG00000213672	skeletal muscle	1.88×10-6 (p-value)	GTEx Portal V6	3
NME6	?	ENSG00000172113	skeletal muscle	2.34×10-6 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	skeletal muscle	5.12×10-10 (p-value)	GTEx Portal V6	3
TMA7	?	ENSG00000232112	skeletal muscle	8.64×10-7 (p-value)	GTEx Portal V6	1
MRPS18AP1	?	ENSG00000229759	prostate	7.45×10-6 (p-value)	GTEx Portal V6	1
MRPS18AP1	?	ENSG00000229759	transverse colon	3.40×10-6 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	transverse colon	2.11×10-6 (p-value)	GTEx Portal V6	1
ZNF589	?	ENSG00000164048	transverse colon	2.46×10-5 (p-value)	GTEx Portal V6	1
MRPS18AP1	?	ENSG00000229759	sun exposed skin	8.13×10-7 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	sun exposed skin	7.28×10-7 (p-value)	GTEx Portal V6	3
NCKIPSD	?	ENSG00000213672	sun exposed skin	1.16×10-5 (p-value)	GTEx Portal V6	1
QRICH1	?	ENSG00000198218	sun exposed skin	2.12×10-5 (p-value)	GTEx Portal V6	1
FCF1P2	?	ENSG00000228638	aorta	1.09×10-6 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	aorta	7.30×10-6 (p-value)	GTEx Portal V6	1
NCKIPSD	?	ENSG00000213672	subcutaneous adipocytes	4.90×10-10 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	subcutaneous adipocytes	5.86×10-9 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	subcutaneous adipocytes	8.77×10-6 (p-value)	GTEx Portal V6	2
QRICH1	?	ENSG00000198218	subcutaneous adipocytes	2.87×10-7 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	subcutaneous adipocytes	6.18×10-8 (p-value)	GTEx Portal V6	3
NME6	?	ENSG00000172113	subcutaneous adipocytes	6.66×10-6 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	visceral adipocytes	1.14×10-6 (p-value)	GTEx Portal V6	3
NME6	?	ENSG00000172113	visceral adipocytes	8.33×10-7 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	spleen	1.08×10-5 (p-value)	GTEx Portal V6	1
NCKIPSD	?	ENSG00000213672	tibial nerve	8.80×10-8 (p-value)	GTEx Portal V6	3
MRPS18AP1	?	ENSG00000229759	tibial nerve	1.33×10-9 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	tibial nerve	1.67×10-10 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	tibial nerve	4.95×10-7 (p-value)	GTEx Portal V6	2
ZNF589	?	ENSG00000164048	esophagus mucosa	1.33×10-6 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	esophagus mucosa	5.92×10-6 (p-value)	GTEx Portal V6	1

QRICH1	?	ENSG00000198218	esophagus mucosa	1.60×10-5 (p-value)	GTEx Portal V6	1
ZNF589	?	ENSG00000164048	adrenal gland	4.93×10-7 (p-value)	GTEx Portal V6	3
QRICH1	?	ENSG00000198218	testis	5.34×10-6 (p-value)	GTEx Portal V6	2
MRPS18AP1	?	ENSG00000229759	testis	6.49×10-8 (p-value)	GTEx Portal V6	3
ZNF589	?	ENSG00000164048	testis	7.45×10-7 (p-value)	GTEx Portal V6	3
ZNF589	ENST00000354698	ILMN_1654612	skin	7.37×10-8 (p-value)	MuTHER consortium	2
ZNF589	ENST00000457782					
ZNF589	ENST00000448461					
WDR6	ENST00000615452	ILMN_1669484	adipocyte	4.99×10-9 (p-value)	MuTHER consortium	2
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
ZNF589	ENST00000354698	ILMN_1654819	blood	2.46×10-6 (p-value)	MuTHER consortium	2
ZNF589	ENST00000457782					
ZNF589	ENST00000448461					
SCAP	ENST00000428413	ILMN_1677534	blood	3.37×10-5 (p-value)	MuTHER consortium	1
SCAP	ENST00000545718					
SCAP	ENST00000320017					
SCAP	ENST00000441517					
SCAP	ENST00000265565					
KLHDC8B	ENST00000332780	ILMN_1695246	blood	2.13×10-9 (p-value)	MuTHER consortium	2
KLHDC8B	ENST00000462582					
AMT	ENST00000476226	ILMN_1665331	b-cell	5.98×10-5 (p-value)	Fairfax et al.	1
AMT	ENST00000495436					
AMT	ENST00000473163					
AMT	ENST00000427987					
AMT	ENST00000399379					
AMT	ENST00000395338					
AMT	ENST00000476127					
AMT	ENST00000273588					
AMT	ENST00000465925					
AMT	ENST00000458307					
AMT	ENST00000538581					
WDR6	ENST00000615452	ILMN_1669484	monocyte	6.65×10-9 (p-value)	Fairfax et al.	1
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
ARIH2OS	ENST00000408959	ILMN_3241670	monocyte	4.42×10-4 (p-value)	Fairfax et al.	1
QRICH1	?	ENSG00000198218	unexposed skin	1.51×10-5 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	left ventricle	5.05×10-6 (p-value)	GTEx Portal V6	1
QRICH1	?	ENSG00000198218	left ventricle	2.08×10-5 (p-value)	GTEx Portal V6	1

NME6	ENST00000415053	ILMN_2229032	blood	1.95×10-5 (p-value)	Westra et al.	2
NME6	ENST00000418431					
NME6	ENST00000426689					
NME6	ENST00000442597					
NME6	ENST00000415644					
NME6	ENST00000447314					
NME6	ENST00000456495					
NME6	ENST00000421967					
NME6	ENST00000452211					
NME6	ENST00000444069					
NME6	ENST00000494209					
NME6	ENST00000425930					
WDR6	ENST00000615452	ILMN_1669484	monocyte	2.13×10-25 (p-value)	Zeller et al.	1
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
NCKIPSD	ENST00000294129	ILMN_1736623	monocyte	1.15×10-12 (p-value)	Zeller et al.	1
NCKIPSD	ENST00000416649					
NCKIPSD	ENST00000470006					
MRPS18AP1	?	ENSG00000229759	coronary artery	1.70×10-5 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
NICN1	ENST00000273598	ILMN_1807211	chr3	monocyte	1.15×10-20 (p-value)	Zeller et al.	1
NICN1	ENST00000615713						
AMT	ENST00000273588						
NICN1	ENST00000423832						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000338474	1	ENCP00000036343	MAP4
		ENCP00000036362	FBXW12
		ENCP00000036405	UQCRC1
		ENCP00000036386	SHISA5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363389 (TF binding site)	1	embryonic stem cell (H1ESC)	PolII, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CAMP	downstream gene variant	1528	ENST00000296435	NM_004345.4	ENSP00000296435	1
CDC25A	upstream gene variant	4866	ENST00000459900	?	?	1
CDC25A	downstream gene variant	3310	ENST00000437972	?	ENSP00000404285	1

NDUFB1P1	upstream gene variant	1928	ENST00000435191	?	?	1
RNU7-128P	upstream gene variant	709	ENST00000517247	?	?	1
ZNF589	downstream gene variant	3557	ENST00000354698	NM_016089.2	ENSP00000346729	1
ZNF589	downstream gene variant	3554	ENST00000457782	?	ENSP00000401106	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CDC25A	ENST00000443342	?	ENSP00000416483	1
CDC25A	ENST00000351231	NM_201567.1	ENSP00000343166	1
CDC25A	ENST00000302506	NM_001789.2	ENSP00000303706	1
ZNF589	ENST00000427617	?	ENSP00000392719	1
ZNF589	ENST00000412564	?	ENSP00000404398	1
ZNF589	ENST00000440261	?	ENSP00000408719	1
ZNF589	ENST00000448461	?	ENSP00000404592	1