Locus 91 (rs4688759)

SNiPAcard

Block annotations

Block info

genomic range	chr3:49,597,055-50,125,108
block size	528,054 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.424 [-1.408 – 3.578]	gene(s) hit or close-by	AC139451.2, ACTBP13, AMIGO3, BSN, CAMKV, GMPPB, IP6K1, RBM5, RBM6, RN7SL217P, RNF123, RP13-1056D16.2, TRAIP
phastCons	μ = 0.194 [0 – 1]	eQTL gene(s)	AMT, APEH, C3orf62, CCDC36, CCDC71, HYAL3, MIR5193, NCKIPSD, NICN1, RBM6, RP11-3B7.1, SEMA3F, SLC26A6, TMEM89, UBA7, USP4, WDR6
GERP++	μ = 0.601 [-1.52 – 5.04]	potentially regulated gene(s)	–
CADD score	μ = 5.022 [0.248 – 16.95]	disease gene(s)	AMT, GMPPB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN [...]	OMIM	MIM:615350
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL [...]	OMIM	MIM:615351
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE), TYPE C, [...]	OMIM	MIM:615352
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYEANOMALIES), TYPE A, 14	DECIPHER	MIM:615350
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863
GMPPB	Autosomal recessive limb-girdle muscular dystrophy type 2T	OrphaNet	OrphaNet:363623
GMPPB	Congenital muscular dystrophy with intellectual disability	OrphaNet	OrphaNet:370968
GMPPB	Muscle eye brain disease	OrphaNet	OrphaNet:588
GMPPB	Congenital muscular dystrophy with cerebellar involvement	OrphaNet	OrphaNet:370959

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
RNF123	missense variant	ENST00000327697	NM_022064.3	ENSP00000328287	H/R	cAc/cGc	?	?	1
RNF123	missense variant	ENST00000432042	?	ENSP00000392443	H/R	cAc/cGc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)

WDR6	?	ENSG00000178252	visceral adipocytes	1.17×10-7 (p-value)	GTEx Portal V6	16
NCKIPSD	?	ENSG00000213672	visceral adipocytes	4.77×10-6 (p-value)	GTEx Portal V6	5
WDR6	?	ENSG00000178252	stomach	1.76×10-8 (p-value)	GTEx Portal V6	16
WDR6	?	ENSG00000178252	lung	8.08×10-8 (p-value)	GTEx Portal V6	16
SLC26A6	?	ENSG00000225697	lung	1.05×10-5 (p-value)	GTEx Portal V6	4
WDR6	?	ENSG00000178252	tibial nerve	1.54×10-11 (p-value)	GTEx Portal V6	16
SEMA3F	?	ENSG00000001617	tibial nerve	3.87×10-7 (p-value)	GTEx Portal V6	18
WDR6	?	ENSG00000178252	transformed fibroblasts	2.14×10-12 (p-value)	GTEx Portal V6	16
NCKIPSD	?	ENSG00000213672	transformed fibroblasts	3.63×10-5 (p-value)	GTEx Portal V6	4
CCDC36	?	ENSG00000173421	transformed fibroblasts	4.92×10-5 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	tibial artery	9.89×10-9 (p-value)	GTEx Portal V6	16
TMEM89	?	ENSG00000183396	tibial artery	1.54×10-5 (p-value)	GTEx Portal V6	5
NICN1	?	ENSG00000145029	tibial artery	6.59×10-8 (p-value)	GTEx Portal V6	18
AMT	?	ENSG00000145020	tibial artery	1.71×10-6 (p-value)	GTEx Portal V6	10
WDR6	?	ENSG00000178252	breast	3.36×10-7 (p-value)	GTEx Portal V6	14
WDR6	?	ENSG00000178252	blood	1.65×10-10 (p-value)	GTEx Portal V6	16
WDR6	?	ENSG00000178252	esophagus mucosa	1.92×10-33 (p-value)	GTEx Portal V6	16
NCKIPSD	?	ENSG00000213672	esophagus mucosa	3.59×10-8 (p-value)	GTEx Portal V6	5
HYAL3	?	ENSG00000186792	esophagus mucosa	1.61×10-5 (p-value)	GTEx Portal V6	4
RBM6	?	ENSG00000004534	esophagus mucosa	4.58×10-5 (p-value)	GTEx Portal V6	2
CCDC36	?	ENSG00000173421	testis	1.22×10-7 (p-value)	GTEx Portal V6	15
WDR6	?	ENSG00000178252	skeletal muscle	5.26×10-10 (p-value)	GTEx Portal V6	16
SLC26A6	?	ENSG00000225697	skeletal muscle	2.04×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	unexposed skin	1.92×10-22 (p-value)	GTEx Portal V6	16
WDR6	?	ENSG00000178252	sun exposed skin	2.32×10-45 (p-value)	GTEx Portal V6	16
RBM6	?	ENSG00000004534	sun exposed skin	3.04×10-6 (p-value)	GTEx Portal V6	7
WDR6	?	ENSG00000178252	terminal ileum	9.22×10-7 (p-value)	GTEx Portal V6	15
WDR6	?	ENSG00000178252	subcutaneous adipocytes	2.03×10-19 (p-value)	GTEx Portal V6	16
RBM6	?	ENSG00000004534	subcutaneous adipocytes	4.30×10-5 (p-value)	GTEx Portal V6	3
CCDC36	?	ENSG00000173421	muscularis mucosae	1.11×10-6 (p-value)	GTEx Portal V6	10
WDR6	ENST00000615452	ILMN_1669484	skin	1.08×10-38 (p-value)	MuTHER consortium	2
WDR6	ENST00000452875		adipocyte	7.89×10-35 (p-value)	MuTHER consortium	2
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					

Locus 91 (sentinel SNP rs4688759 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	skin	4.51×10-8 (p-value)	MuTHER consortium	3
USP4	ENST00000483212		adipocyte	5.30×10-13 (p-value)	MuTHER consortium	3
USP4	ENST00000485450		blood	6.42×10-5 (p-value)	MuTHER consortium	3
C3orf62	ENST00000343010					
RP11-3B7.1	ENST00000622888	ILMN_1807244	adipocyte	2.17×10-18 (p-value)	MuTHER consortium	3
RP11-3B7.1	ENST00000440528					
WDR6	ENST00000615452	ILMN_1669484	monocyte	1.33×10-29 (p-value)	Zeller et al.	1
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	monocyte	2.05×10-34 (p-value)	Zeller et al.	1
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
APEH	ENST00000480772	ILMN_1718023	blood	1.03×10-4 (p-value)	Westra et al.	2
APEH	ENST00000469362					
APEH	ENST00000296456					
APEH	ENST00000438011					
WDR6	ENST00000615452	ILMN_1669484	monocyte	3.22×10-14 (p-value)	Fairfax et al.	2
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
UBA7	ENST00000497908	ILMN_1794612	b-cell	3.40×10-5 (p-value)	Fairfax et al.	2
UBA7	ENST00000333486					
MIR5193	ENST00000584510					
USP4	ENST00000351842	ILMN_1798712	monocyte	6.64×10-11 (p-value)	Fairfax et al.	2
USP4	ENST00000483212		b-cell	5.42×10-8 (p-value)	Fairfax et al.	2
C3orf62	ENST00000343010					
USP4	ENST00000485450					
CCDC71	ENST00000321895	ILMN_1768433	monocyte	1.86×10-4 (p-value)	Fairfax et al.	1
			b-cell	6.44×10-4 (p-value)	Fairfax et al.	1
SLC26A6	ENST00000420764	ILMN_1785252	b-cell	5.46×10-6 (p-value)	Fairfax et al.	2
SLC26A6	ENST00000395550					
SLC26A6	ENST00000489483					
SLC26A6	ENST00000480524					
SLC26A6	ENST00000455886					
SLC26A6	ENST00000307364					
SLC26A6	ENST00000614797					
SLC26A6	ENST00000358747					
SLC26A6	ENST00000383733					
SLC26A6	ENST00000337000					
RBM6	ENST00000442092	ILMN_1738239	monocyte	2.69×10-4 (p-value)	Fairfax et al.	1

RBM6	ENST00000266022		b-cell	1.61×10-4 (p-value)	Fairfax et al.	1
RBM6	ENST00000454079					
RBM6	ENST00000422955					
RBM6	ENST00000419610					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
HYAL3	?	ENSG00000186792	left ventricle	2.01×10-6 (p-value)	GTEx Portal V6	5

Putative effect on regulation

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
RNF123	1	ENST00000327697 ENST00000432042 ENST00000457726 ENST00000486102 ENST00000487805	hsa-miR-504-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC139451.2	upstream gene variant	2051	ENST00000627736	?	?	1
ACTBP13	downstream gene variant	2442	ENST00000423971	?	?	1
AMIGO3	downstream gene variant	4291	ENST00000320431	NM_198722.2	ENSP00000323096	1
CAMKV	upstream gene variant	1000	ENST00000466535	?	ENSP00000420766	1
CAMKV	upstream gene variant	683	ENST00000477224	NM_024046.3	ENSP00000419195	1
CAMKV	upstream gene variant	974	ENST00000463537	?	ENSP00000417614	1
CAMKV	upstream gene variant	1000	ENST00000498324	?	?	1
CAMKV	upstream gene variant	4491	ENST00000478149	?	?	1
CAMKV	upstream gene variant	970	ENST00000480398	?	ENSP00000420000	1
CAMKV	upstream gene variant	3109	ENST00000483811	?	?	1
CAMKV	upstream gene variant	1000	ENST00000296471	?	ENSP00000296471	1
CAMKV	upstream gene variant	999	ENST00000467248	?	ENSP00000420053	1
CAMKV	upstream gene variant	1000	ENST00000479704	?	?	1
CAMKV	upstream gene variant	996	ENST00000466940	?	ENSP00000420724	1
CAMKV	upstream gene variant	1000	ENST00000476105	?	?	1
CAMKV	upstream gene variant	1002	ENST00000475665	?	?	1
CAMKV	upstream gene variant	3127	ENST00000620470	?	ENSP00000484045	1
CAMKV	upstream gene variant	999	ENST00000488336	?	ENSP00000418809	1
CAMKV	upstream gene variant	3109	ENST00000472895	?	?	1
CAMKV	upstream gene variant	1002	ENST00000487726	?	ENSP00000420139	1
GMPPB	downstream gene variant	4301	ENST00000480687	?	ENSP00000418565	1
RBM5	upstream gene variant	1279	ENST00000441305	?	ENSP00000390711	1
RBM5	upstream gene variant	2695	ENST00000417905	?	ENSP00000406119	1
RBM5	upstream gene variant	1244	ENST00000404526	?	ENSP00000384872	1
RBM5	upstream gene variant	2206	ENST00000437500	?	ENSP00000394622	1
RBM5	upstream gene variant	1280	ENST00000438369	?	ENSP00000414218	1

RBM5	upstream gene variant	1279	ENST00000433556	?	?	1
RBM5	upstream gene variant	1244	ENST00000469838	?	ENSP00000419534	1
RBM5	upstream gene variant	1233	ENST00000347869	NM_005778.3	ENSP00000343054	1
RBM5	upstream gene variant	1300	ENST00000492472	?	?	1
RBM5	upstream gene variant	1288	ENST00000464087	?	?	1
RBM6	downstream gene variant	3031	ENST00000433811	?	ENSP00000389763	1
RBM6	downstream gene variant	933	ENST00000493652	?	?	1
RBM6	downstream gene variant	3327	ENST00000491874	?	?	1
RBM6	downstream gene variant	3320	ENST00000488807	?	?	1
RBM6	downstream gene variant	1423	ENST00000438912	?	ENSP00000403865	1
RBM6	downstream gene variant	42	ENST00000441115	?	?	1
RN7SL217P	upstream gene variant	1855	ENST00000584520	?	?	1
RNF123	upstream gene variant	287	ENST00000433785	?	ENSP00000416156	1
RNF123	upstream gene variant	984	ENST00000444689	?	ENSP00000413794	1
RNF123	upstream gene variant	7	ENST00000469978	?	?	1
RP13-1056D16.2	downstream gene variant	211	ENST00000463176	?	?	2
TRAIP	downstream gene variant	2334	ENST00000488860	?	?	2
TRAIP	downstream gene variant	2344	ENST00000482243	?	ENSP00000419350	2
TRAIP	downstream gene variant	3564	ENST00000489948	?	?	1
TRAIP	downstream gene variant	3699	ENST00000473863	?	?	1
TRAIP	downstream gene variant	2993	ENST00000477546	?	?	1
TRAIP	downstream gene variant	1915	ENST00000482582	?	ENSP00000418544	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
RBM6	ENST00000443081	?	ENSP00000396466	Q	caA/caG	1
RBM6	ENST00000442092	NM_001167582.1	ENSP00000393530	Q	caA/caG	1
RBM6	ENST00000266022	NM_005777.2	ENSP00000266022	Q	caA/caG	1
RBM6	ENST00000422955	?	ENSP00000392939	Q	caA/caG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BSN	ENST00000296452	NM_003458.3	ENSP00000296452	5
CAMKV	ENST00000463537	?	ENSP00000417614	1
CAMKV	ENST00000477224	NM_024046.3	ENSP00000419195	1
CAMKV	ENST00000296471	?	ENSP00000296471	1
CAMKV	ENST00000466535	?	ENSP00000420766	1
CAMKV	ENST00000487726	?	ENSP00000420139	1
CAMKV	ENST00000475665	?	?	1
CAMKV	ENST00000467248	?	ENSP00000420053	1

CAMKV	ENST00000476105	?	?	1
CAMKV	ENST00000480398	?	ENSP00000420000	1
CAMKV	ENST00000479704	?	?	1
CAMKV	ENST00000466940	?	ENSP00000420724	1
CAMKV	ENST00000488336	?	ENSP00000418809	1
CAMKV	ENST00000498324	?	?	1
IP6K1	ENST00000613416	NM_001242829.1	ENSP00000482032	2
IP6K1	ENST00000321599	NM_153273.3	ENSP00000323780	2
IP6K1	ENST00000395238	NM_001006115.2	ENSP00000378659	2
IP6K1	ENST00000468463	?	ENSP00000420467	2
IP6K1	ENST00000498149	?	?	2
IP6K1	ENST00000460540	?	ENSP00000420762	2
RBM6	ENST00000483350	?	?	2
RBM6	ENST00000493652	?	?	2
RBM6	ENST00000441115	?	?	6
RBM6	ENST00000443081	?	ENSP00000396466	6
RBM6	ENST00000464013	?	?	2
RBM6	ENST00000434592	?	ENSP00000399942	7
RBM6	ENST00000488807	?	?	3
RBM6	ENST00000442092	NM_001167582.1	ENSP00000393530	6
RBM6	ENST00000491874	?	?	3
RBM6	ENST00000425608	?	ENSP00000408665	6
RBM6	ENST00000266022	NM_005777.2	ENSP00000266022	6
RBM6	ENST00000454079	?	ENSP00000406548	6
RBM6	ENST00000422955	?	ENSP00000392939	6
RBM6	ENST00000419610	?	ENSP00000396410	6
RBM6	ENST00000433811	?	ENSP00000389763	3
TRAIP	ENST00000475495	?	?	2
TRAIP	ENST00000473195	?	ENSP00000419556	2
TRAIP	ENST00000482582	?	ENSP00000418544	1
TRAIP	ENST00000331456	NM_005879.2	ENSP00000328203	2
TRAIP	ENST00000469027	?	ENSP00000420085	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RBM6	ENST00000434592	?	ENSP00000399942	1
RBM6	ENST00000454079	?	ENSP00000406548	1
RBM6	ENST00000419610	?	ENSP00000396410	1
RNF123	ENST00000457726	?	ENSP00000394369	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RBM6	ENST00000421682	?	ENSP00000392475	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RNF123	ENST00000487805	?	1
RNF123	ENST00000486102	?	1