Locus 82 (rs2239908)

SNiPAcard

Block annotations

Block info

genomic range	chr17:26,703,682-26,740,139
block size	36,458 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.006 [-1.561 – 1.574]	gene(s) hit or close-by	CTB-96E2.10, CTB-96E2.3, CTB-96E2.6, CTD-2350C19.2, H3F3BP2, SARM1, SLC46A1, VTN
phastCons	μ = 0.067 [0 – 0.654]	eQTL gene(s)	CTB-96E2.3, CTB-96E2.7, POLDIP2, SARM1, TMEM199, TMEM97
GERP++	μ = -0.561 [-6.27 – 2.35]	potentially regulated gene(s)	–
CADD score	μ = 5.301 [0.45 – 10.68]	disease gene(s)	SLC46A1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SLC46A1	hereditary folate malabsorption (HFM)	DECIPHER	MIM:229050
SLC46A1	FOLATE MALABSORPTION, HEREDITARY	OMIM	MIM:229050
SLC46A1	Hereditary folate malabsorption	OrphaNet	OrphaNet:90045

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TMEM199	?	ENSG00000244045	muscularis mucosae	1.58×10^{-7} (p-value)	GTEx Portal V6	15
POLDIP2	?	ENSG00000004142	nucleus accumbens	10.00×10^{-7} (p-value)	GTEx Portal V6	6
TMEM199	?	ENSG00000244045	lung	6.65×10^{-7} (p-value)	GTEx Portal V6	7
CTB-96E2.3	?	ENSG00000258924	lung	1.90×10^{-8} (p-value)	GTEx Portal V6	17
POLDIP2	?	ENSG00000004142	cortex	1.22×10^{-6} (p-value)	GTEx Portal V6	9
CTB-96E2.7	?	ENSG00000265618	tibial nerve	1.77×10^{-5} (p-value)	GTEx Portal V6	6
TMEM199	?	ENSG00000244045	tibial nerve	1.23×10^{-12} (p-value)	GTEx Portal V6	17
POLDIP2	?	ENSG00000004142	tibial nerve	2.60×10^{-10} (p-value)	GTEx Portal V6	18
TMEM199	?	ENSG00000244045	anterior cingulate cortex	2.17×10^{-6} (p-value)	GTEx Portal V6	1
POLDIP2	?	ENSG00000004142	putamen	7.53×10^{-6} (p-value)	GTEx Portal V6	2
TMEM199	?	ENSG00000244045	unexposed skin	4.06×10^{-6} (p-value)	GTEx Portal V6	6
TMEM199	?	ENSG00000244045	sun exposed skin	4.21×10^{-6} (p-value)	GTEx Portal V6	2
TMEM199	?	ENSG00000244045	subcutaneous adipocytes	9.44×10^{-8} (p-value)	GTEx Portal V6	11
POLDIP2	?	ENSG00000004142	subcutaneous adipocytes	1.92×10^{-5} (p-value)	GTEx Portal V6	6

TMEM199	?	ENSG00000244045	EBV lymphocytes	6.74×10-6 (p-value)	GTEx Portal V6	1
SARM1	ENST00000585482	ILMN_1746265	skin	2.09×10-6 (p-value)	MuTHER consortium	7
			blood	5.04×10-12 (p-value)	MuTHER consortium	7
			adipocyte	1.76×10-12 (p-value)	MuTHER consortium	7
TMEM97	ENST00000226230	ILMN_1710962	blood	1.67×10-14 (p-value)	MuTHER consortium	7
TMEM97	ENST00000336687		adipocyte	2.30×10-14 (p-value)	MuTHER consortium	7
TMEM97	ENST00000582384					
POLDIP2	ENST00000540200	ILMN_1743499	blood	7.99×10-5 (p-value)	Westra et al.	1
POLDIP2	?	ENSG00000004142	testis	9.09×10-6 (p-value)	GTEx Portal V6	4
TMEM97	?	ENSG00000109084	blood	2.78×10-3 (q-value)	SeeQTL DB (HapMap)	1
SARM1	ENST00000585482	ILMN_1746265	monocyte	1.61×10-16 (p-value)	Zeller et al.	1
TMEM97	ENST00000226230	ILMN_1710962	monocyte	4.23×10-36 (p-value)	Zeller et al.	1
TMEM97	ENST00000336687					
TMEM97	ENST00000582384					

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
MAX	MA0058.2	6	yes	-0.145	1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001343354 (promoter flanking region)	3	cervix (HeLa-S3)	DNase1, H3K4me1
		NHLF	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun
		liver (HepG2)	PolII
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00001343356 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K27me3
		liver (HepG2)	PolII, H3K4me1
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTB-96E2.10	downstream gene variant	3101	ENST00000613598	?	?	1
CTB-96E2.6	upstream gene variant	2362	ENST00000623773	?	?	1
CTD-2350C19.2	downstream gene variant	4074	ENST00000580714	?	?	1
H3F3BP2	upstream gene variant	3801	ENST00000583907	?	?	1
SARM1	downstream gene variant	1408	ENST00000582323	?	ENSP00000464166	5
SARM1	downstream gene variant	396	ENST00000577870	?	?	4
SARM1	downstream gene variant	86	ENST00000585453	?	?	2
SARM1	downstream gene variant	183	ENST00000578128	?	ENSP00000462479	4
SARM1	downstream gene variant	616	ENST00000580711	?	?	4

SARM1	downstream gene variant	616	ENST00000580711	?	?	4
SARM1	downstream gene variant	2631	ENST00000585482	NM_015077.3	ENSP00000468032	1
SARM1	downstream gene variant	1429	ENST00000579593	?	ENSP00000462228	2
SARM1	downstream gene variant	114	ENST00000379061	?	?	2
SLC46A1	downstream gene variant, upstream gene variant	470	ENST00000612814	NM_080669.5	ENSP00000480703	9
SLC46A1	upstream gene variant	50	ENST00000584995	?	ENSP00000464190	2
SLC46A1	upstream gene variant	933	ENST00000578217	?	?	1
SLC46A1	downstream gene variant, upstream gene variant	50	ENST00000618626	NM_001242366.2	ENSP00000483652	9
SLC46A1	downstream gene variant, upstream gene variant	618	ENST00000582735	?	ENSP00000463339	9
SLC46A1	upstream gene variant	81	ENST00000584426	?	ENSP00000467416	2
SLC46A1	downstream gene variant	1790	ENST00000619923	?	?	6
SLC46A1	upstream gene variant	3673	ENST00000581516	?	ENSP00000462942	1
SLC46A1	upstream gene variant	512	ENST00000582590	?	?	2
VTN	upstream gene variant	3572	ENST00000542029	?	ENSP00000440439	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTB-96E2.3	ENST00000591482	?	?	1
SARM1	ENST00000585453	?	?	13
SARM1	ENST00000578128	?	ENSP00000462479	11
SARM1	ENST00000585482	NM_015077.3	ENSP00000468032	14
SARM1	ENST00000379061	?	?	14
SARM1	ENST00000579593	?	ENSP00000462228	13
SARM1	ENST00000577870	?	?	11
SARM1	ENST00000580711	?	?	11
SLC46A1	ENST00000581516	?	ENSP00000462942	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SARM1	ENST00000585482	NM_015077.3	ENSP00000468032	8
SARM1	ENST00000579593	?	ENSP00000462228	6
SLC46A1	ENST00000618626	NM_001242366.2	ENSP00000483652	5
SLC46A1	ENST00000612814	NM_080669.5	ENSP00000480703	8

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTD-2350C19.2	ENST00000580714	?	1
SARM1	ENST00000585453	?	2