Locus 312 (rs2236293)

SNiPAcard

Block annotations

Block info

genomic range	chr9:35,779,571-35,860,942
block size	81,372 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.196 [-1.18 – 3.035]	gene(s) hit or close-by	FAM221B, HINT2, LINC00950, NPR2, OR13E1P, RP11-112J3.16, SPAG8, TMEM8B
phastCons	μ = 0.072 [0 – 0.999]	eQTL gene(s)	GBA2, HRCT1, MSMP, NPR2, RP11-112J3.16, RP11-182N22.8, SPAG8, TMEM8B, TPM2
GERP++	μ = 0.459 [-5.31 – 4.04]	potentially regulated gene(s)	–
CADD score	μ = 6.165 [0.737 – 17.84]	disease gene(s)	NPR2, GBA2, TPM2

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
2-aminobutyrate	5.07×10^{-5}	Metabolomics GWAS Server	24816252	3

Disease gene annotation

gene	trait	source DB	source entry/link
NPR2	acromesomelic dysplasia Maroteaux type (AMDM)	DECIPHER	MIM:602875
NPR2	ACROMESOMELIC DYSPLASIA, MAROTEAUX TYPE	OMIM	MIM:602875
NPR2	EPIPHYSEAL CHONDRODYSPLASIA, MIURA TYPE	OMIM	MIM:615923
GBA2	SPASTIC PARAPLEGIA 46, AUTOSOMAL RECESSIVE	OMIM	MIM:614409
TPM2	ARTHROGRYPOSIS, DISTAL, TYPE 1A	OMIM	MIM:108120
TPM2	ARTHROGRYPOSIS, DISTAL, TYPE 2B	OMIM	MIM:601680
TPM2	NEMALINE MYOPATHY 4	OMIM	MIM:609285
NPR2	ACROMESOMELIC DYSPLASIA, MAROTEAUX TYPE	OrphaNet	OrphaNet:40
NPR2	Tall stature - scoliosis - macrodactyly of the great toes	OrphaNet	OrphaNet:329191
GBA2	Autosomal recessive cerebellar ataxia with late-onset spasticity	OrphaNet	OrphaNet:352641
GBA2	Autosomal recessive spastic paraplegia type 46	OrphaNet	OrphaNet:320391
TPM2	Sheldon-Hall syndrome	OrphaNet	OrphaNet:1147
TPM2	Congenital fiber-type disproportion myopathy	OrphaNet	OrphaNet:2020
TPM2	Trismus - pseudocamptodactyly	OrphaNet	OrphaNet:3377
TPM2	Cap myopathy	OrphaNet	OrphaNet:171881
TPM2	Typical nemaline myopathy	OrphaNet	OrphaNet:171436
TPM2	Childhood-onset nemaline myopathy	OrphaNet	OrphaNet:171439
TPM2	Digitotalar dysmorphism	OrphaNet	OrphaNet:1146

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HRCT1	ENST00000354323	ILMN_2189675	adipocyte	6.09×10^{-8} (p-value)	MuTHER consortium	5
MSMP	ENST00000436428	ILMN_2115135	blood	1.08×10^{-7} (p-value)	MuTHER consortium	10

MSMP	ENST00000414286					
NPR2	ENST00000421267	ILMN_1681994	skin	1.81×10-5 (p-value)	MuTHER consortium	5
NPR2	ENST00000469249					
NPR2	ENST00000464810					
TMEM8B	?	ENSG00000137103	muscularis mucosae	3.39×10-8 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	lung	1.62×10-10 (p-value)	GTEx Portal V6	17
GBA2	?	ENSG00000070610	atrial appendage	6.96×10-8 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	atrial appendage	8.94×10-7 (p-value)	GTEx Portal V6	5
TMEM8B	?	ENSG00000137103	transformed fibroblasts	8.32×10-10 (p-value)	GTEx Portal V6	17
GBA2	?	ENSG00000070610	blood	1.72×10-8 (p-value)	GTEx Portal V6	14
TMEM8B	?	ENSG00000137103	breast	9.66×10-6 (p-value)	GTEx Portal V6	2
TMEM8B	?	ENSG00000137103	tibial artery	7.01×10-14 (p-value)	GTEx Portal V6	17
NPR2	?	ENSG00000159899	tibial artery	2.55×10-6 (p-value)	GTEx Portal V6	7
MSMP	ENST00000436428	ILMN_2115135	b-cell	5.37×10-7 (p-value)	Fairfax et al.	7
MSMP	ENST00000414286					
GBA2	ENST00000486797	ILMN_1674560	monocyte	1.95×10-5 (p-value)	Fairfax et al.	1
GBA2	ENST00000378103					
GBA2	ENST00000467252					
GBA2	ENST00000378088					
GBA2	ENST00000378094					
NPR2	?	ENSG00000159899	thyroid	1.02×10-5 (p-value)	GTEx Portal V6	4
TMEM8B	?	ENSG00000137103	thyroid	5.05×10-10 (p-value)	GTEx Portal V6	17
RP11-182N22.8	?	ENSG00000234181	thyroid	1.38×10-5 (p-value)	GTEx Portal V6	2
GBA2	?	ENSG00000070610	skeletal muscle	2.77×10-12 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	skeletal muscle	1.27×10-19 (p-value)	GTEx Portal V6	17
RP11-112J3.16	?	ENSG00000227388	skeletal muscle	1.70×10-5 (p-value)	GTEx Portal V6	1
TMEM8B	?	ENSG00000137103	sun exposed skin	6.07×10-11 (p-value)	GTEx Portal V6	17
HRCT1	?	ENSG00000196196	sun exposed skin	1.30×10-5 (p-value)	GTEx Portal V6	1
TMEM8B	?	ENSG00000137103	unexposed skin	8.29×10-9 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	aorta	4.86×10-12 (p-value)	GTEx Portal V6	17
NPR2	?	ENSG00000159899	left ventricle	4.74×10-7 (p-value)	GTEx Portal V6	7
GBA2	?	ENSG00000070610	left ventricle	2.30×10-11 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	left ventricle	1.06×10-7 (p-value)	GTEx Portal V6	8
TMEM8B	?	ENSG00000137103	subcutaneous adipocytes	1.42×10-7 (p-value)	GTEx Portal V6	17
SPAG8	ENST00000489063	ILMN_1754370	monocyte	2.27×10-18 (p-value)	Zeller et al.	4
SPAG8	ENST00000340291					
SPAG8	ENST00000475644					
SPAG8	ENST00000460836					
SPAG8	ENST00000463889					
TMEM8B	?	ENSG00000137103	coronary artery	1.67×10-8 (p-value)	GTEx Portal V6	17

NPR2	?	ENSG00000159899	tibial nerve	2.31×10-6 (p-value)	GTEx Portal V6	9
GBA2	?	ENSG00000070610	tibial nerve	1.01×10-6 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	tibial nerve	1.58×10-14 (p-value)	GTEx Portal V6	17
TMEM8B	?	ENSG00000137103	esophagus mucosa	5.83×10-11 (p-value)	GTEx Portal V6	17
HRCT1	?	ENSG00000196196	esophagus mucosa	3.92×10-6 (p-value)	GTEx Portal V6	3
NPR2	?	ENSG00000159899	adrenal gland	4.43×10-8 (p-value)	GTEx Portal V6	14
TMEM8B	?	ENSG00000137103	adrenal gland	1.58×10-7 (p-value)	GTEx Portal V6	9
SPAG8	ENST00000489063	ILMN_1754370	blood	7.98×10-5 (p-value)	Westra et al.	1
SPAG8	ENST00000340291					
SPAG8	ENST00000475644					
SPAG8	ENST00000460836					
SPAG8	ENST00000463889					
TPM2	ENST00000378300	ILMN_1789196	blood	1.10×10-6 (p-value)	Westra et al.	6
TPM2	ENST00000329305					
TPM2	ENST00000378292					
TMEM8B	?	ENSG00000137103	stomach	4.38×10-7 (p-value)	GTEx Portal V6	8
TMEM8B	?	ENSG00000137103	uterus	2.09×10-7 (p-value)	GTEx Portal V6	7

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001470336 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	Ini1, DNase1, Max, USF1
		breast (HMEC)	DNase1
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3
		endothelium (HUVEC)	Max, DNase1, Cjun
		lung (IMR90)	DNase1, H3K36me3
		A549	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FAM221B	downstream gene variant	1762	ENST00000377984	?	ENSP00000367222	1
HINT2	upstream gene variant	2561	ENST00000259667	NM_032593.2	ENSP00000259667	1
HINT2	upstream gene variant	3888	ENST00000471774	?	?	1
HINT2	upstream gene variant	2590	ENST00000472085	?	?	1
HINT2	upstream gene variant	2228	ENST00000474848	?	?	1
HINT2	upstream gene variant	2230	ENST00000474908	?	?	1
HINT2	upstream gene variant	2561	ENST00000461169	?	?	1
HINT2	upstream gene variant	3812	ENST00000490578	?	?	1
LINC00950	upstream gene variant	1111	ENST00000629651	?	?	1
NPR2	upstream gene variant	1933	ENST00000447210	?	ENSP00000393029	1

NPR2	upstream gene variant	4249	ENST00000469249	?	?	1
NPR2	upstream gene variant	1378	ENST00000421267	?	ENSP00000399204	1
NPR2	upstream gene variant	2381	ENST00000448821	?	ENSP00000402902	1
OR13E1P	downstream gene variant, upstream gene variant	503	ENST00000417104	?	?	3
RP11-112J3.16	downstream gene variant	1871	ENST00000574939	?	?	1
RP11-112J3.16	downstream gene variant	4484	ENST00000431981	?	?	1
SPAG8	downstream gene variant	3893	ENST00000463889	?	?	1
SPAG8	downstream gene variant	3895	ENST00000475644	?	ENSP00000418530	1
SPAG8	downstream gene variant	3893	ENST00000340291	NM_172312.1	ENSP00000340982	1
SPAG8	downstream gene variant	3979	ENST00000489063	?	?	1
SPAG8	downstream gene variant	3893	ENST00000460836	?	?	1
TMEM8B	downstream gene variant	2781	ENST00000377988	NM_001042590.2	ENSP00000367227	1
TMEM8B	downstream gene variant	525	ENST00000472010	?	?	2
TMEM8B	downstream gene variant	2780	ENST00000377991	NM_001042589.2	ENSP00000367230	1
TMEM8B	downstream gene variant	390	ENST00000464519	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NPR2	ENST00000464810	?	?	3
NPR2	ENST00000342694	NM_003995.3	ENSP00000341083	3
RP11-112J3.16	ENST00000431981	?	?	3
RP11-112J3.16	ENST00000574939	?	?	1
TMEM8B	ENST00000472010	?	?	5
TMEM8B	ENST00000377988	NM_001042590.2	ENSP00000367227	7
TMEM8B	ENST00000377996	?	ENSP00000367235	8
TMEM8B	ENST00000439587	NM_016446.3	ENSP00000395810	7
TMEM8B	ENST00000464519	?	?	6
TMEM8B	ENST00000473947	?	?	7
TMEM8B	ENST00000377991	NM_001042589.2	ENSP00000367230	7
TMEM8B	ENST00000490199	?	?	7

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FAM221B	ENST00000388950	?	ENSP00000373602	1
FAM221B	ENST00000423537	NM_001012446.3	ENSP00000415299	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TMEM8B	ENST00000377988	NM_001042590.2	ENSP00000367227	1
TMEM8B	ENST00000377996	?	ENSP00000367235	1
TMEM8B	ENST00000439587	NM_016446.3	ENSP00000395810	1

TMEM8B	ENST00000377991	NM_001042589.2	ENSP00000367230	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
LINC00950	ENST00000629651	?	2
TMEM8B	ENST00000464519	?	1