Locus 237 (rs1005776)

SNiPAcard

Block annotations

Block info

genomic range	chr14:102,807,088-102,904,179
block size	97,092 bp
variant count	21 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.144 [-4.741 – 1.045]	gene(s) hit or close-by	AL137229.1, CINP, TECPR2, ZNF839
phastCons	μ = 0.025 [0 – 0.295]	eQTL gene(s)	ANKRD9, CINP, TECPR2, ZNF839
GERP++	μ = -0.697 [-5.15 – 2.34]	potentially regulated gene(s)	–
CADD score	μ = 2.528 [0.153 – 6.923]	disease gene(s)	TECPR2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TECPR2	SPASTIC PARAPLEGIA 49, AUTOSOMAL RECESSIVE	OMIM	MIM:615031
TECPR2	Hereditary Spastic Paraparesis	DECIPHER	MIM:615031
TECPR2	Autosomal recessive spastic paraplegia type 49	OrphaNet	OrphaNet:320385

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CINP	?	ENSG00000100865	pancreas	2.73×10^{-6} (p-value)	GTEx Portal V6	4
CINP	?	ENSG00000100865	muscularis mucosae	1.08×10^{-13} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	lung	7.13×10^{-13} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	atrial appendage	1.82×10^{-9} (p-value)	GTEx Portal V6	16
ANKRD9	?	ENSG00000156381	transformed fibroblasts	3.75×10^{-25} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	transformed fibroblasts	4.66×10^{-15} (p-value)	GTEx Portal V6	20
TECPR2	?	ENSG00000196663	tibial artery	6.53×10^{-7} (p-value)	GTEx Portal V6	15
CINP	?	ENSG00000100865	tibial artery	1.15×10^{-10} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	breast	1.45×10^{-10} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	blood	8.51×10^{-10} (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	thyroid	9.94×10^{-12} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	skeletal muscle	1.51×10^{-15} (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	transverse colon	1.62×10^{-9} (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	EBV lymphocytes	7.87×10^{-9} (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	unexposed skin	2.69×10^{-11} (p-value)	GTEx Portal V6	20

CINP	?	ENSG00000100865	sun exposed skin	8.24×10-17 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	liver	4.17×10-9 (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	left ventricle	1.20×10-10 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	aorta	1.49×10-11 (p-value)	GTEx Portal V6	19
CINP	?	ENSG00000100865	subcutaneous adipocytes	2.11×10-16 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	visceral adipocytes	5.94×10-14 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	tibial nerve	2.58×10-11 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	coronary artery	8.52×10-8 (p-value)	GTEx Portal V6	8
CINP	?	ENSG00000100865	sigmoid colon	5.46×10-9 (p-value)	GTEx Portal V6	20
CINP	?	ENSG00000100865	esophagus mucosa	1.18×10-15 (p-value)	GTEx Portal V6	20
CINP	ENST00000216756	ILMN_1765257	blood	6.74×10-6 (p-value)	Westra et al.	8
CINP	ENST00000541568					
CINP	ENST00000559504					
CINP	ENST00000559514					
CINP	ENST00000536961					
ANKRD9	ENST00000559404	ILMN_1675937	blood	5.57×10-7 (p-value)	Westra et al.	5
ANKRD9	ENST00000286918					
ANKRD9	ENST00000559651					
ANKRD9	ENST00000557902					
ANKRD9	ENST00000560748					
CINP	?	ENSG00000100865	gastroesophageal junction	3.54×10-9 (p-value)	GTEx Portal V6	18
CINP	?	ENSG00000100865	testis	1.38×10-8 (p-value)	GTEx Portal V6	19
ZNF839	ENST00000558490	ILMN_2186858	skin	3.87×10-9 (p-value)	MuTHER consortium	8
ZNF839	ENST00000420933		adipocyte	2.40×10-7 (p-value)	MuTHER consortium	8
ZNF839	ENST00000557803		blood	3.39×10-5 (p-value)	MuTHER consortium	8
ZNF839	ENST00000559098					
ZNF839	ENST00000561251					
ZNF839	ENST00000558850					
ZNF839	ENST00000559155					
ZNF839	ENST00000559185					
ZNF839	ENST00000442396					
ZNF839	ENST00000560112					
ZNF839	ENST00000558462					
ZNF839	ENST00000420933	ILMN_2186858	b-cell	1.12×10-6 (p-value)	Fairfax et al.	2
ZNF839	ENST00000558490					
ZNF839	ENST00000557803					
ZNF839	ENST00000559098					
ZNF839	ENST00000559155					
ZNF839	ENST00000558850					
ZNF839	ENST00000561251					
ZNF839	ENST00000559185					
ZNF839	ENST00000442396					
ZNF839	ENST00000560112					
ZNF839	ENST00000558462					
CINP	?	ENSG00000100865	adrenal gland	4.59×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000424047 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27ac, H3K36me3, DNase1
ENSR00001625363 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K36me3
		blood (K562)	H3K27me3
ENSR00000424048 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, CTCF, H2AZ, Rad21, Egr1
		skin (NHDF-AD)	CTCF, H3K4me2, DNase1, H3K4me3
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, CTCF, Rad21
		blood (GM12878)	CTCF, DNase1, H2AZ, Yy1, Rad21
		lung (IMR90)	H3K4me2, H3K36me3, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K36me3, CTCF, H3K4me2
		blood (DND-41)	CTCF, H3K36me3
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		A549	H3K4me3, CTCF, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL137229.1	upstream gene variant, downstream gene variant	218	ENST00000631034	?	?	3
CINP	downstream gene variant	351	ENST00000559514	?	ENSP00000453839	1
CINP	downstream gene variant	1868	ENST00000559504	?	ENSP00000453846	1
CINP	downstream gene variant, upstream gene variant	2746	ENST00000560326	?	?	3
CINP	downstream gene variant	1712	ENST00000558523	?	?	2
CINP	downstream gene variant	955	ENST00000536961	?	ENSP00000442057	2
CINP	downstream gene variant	2389	ENST00000561468	?	?	1
CINP	downstream gene variant	752	ENST00000216756	NM_032630.2	ENSP00000216756	2
CINP	downstream gene variant	2476	ENST00000558764	?	ENSP00000452982	1
CINP	downstream gene variant	981	ENST00000541568	?	ENSP00000442377	2
TECPR2	downstream gene variant	1663	ENST00000561228	?	?	1
TECPR2	upstream gene variant	4167	ENST00000560060	?	?	2
TECPR2	upstream gene variant	435	ENST00000557786	?	?	1
ZNF839	downstream gene variant	858	ENST00000558850	NM_001267827.1	ENSP00000453363	2
ZNF839	downstream gene variant	520	ENST00000561251	?	ENSP00000453137	3
ZNF839	downstream gene variant	855	ENST00000557803	?	?	2
ZNF839	downstream gene variant	855	ENST00000559155	?	ENSP00000453415	2
ZNF839	downstream gene variant	1246	ENST00000560112	?	ENSP00000453744	2
ZNF839	downstream gene variant	855	ENST00000559098	?	ENSP00000453515	2
ZNF839	downstream gene variant	858	ENST00000558462	?	ENSP00000454159	2

ZNF839	downstream gene variant	855	ENST00000420933	?	?	2
ZNF839	downstream gene variant	1921	ENST00000560568	?	ENSP00000452983	2
ZNF839	downstream gene variant	855	ENST00000558490	?	ENSP00000452819	2
ZNF839	downstream gene variant	855	ENST00000442396	NM_018335.4	ENSP00000399863	2
ZNF839	downstream gene variant	855	ENST00000559185	NM_001267828.1	ENSP00000453109	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CINP	ENST00000559504	?	ENSP00000453846	4
CINP	ENST00000559514	?	ENSP00000453839	4
CINP	ENST00000536961	?	ENSP00000442057	2
CINP	ENST00000216756	NM_032630.2	ENSP00000216756	2
CINP	ENST00000561468	?	?	2
CINP	ENST00000541568	?	ENSP00000442377	2
CINP	ENST00000558764	?	ENSP00000452982	2
CINP	ENST00000560326	?	?	1
TECPR2	ENST00000558678	NM_001172631.1	ENSP00000453671	15
TECPR2	ENST00000359520	NM_014844.3	ENSP00000352510	15
TECPR2	ENST00000560060	?	?	1
TECPR2	ENST00000561228	?	?	11
ZNF839	ENST00000558850	NM_001267827.1	ENSP00000453363	1
ZNF839	ENST00000559098	?	ENSP00000453515	1
ZNF839	ENST00000560112	?	ENSP00000453744	1
ZNF839	ENST00000442396	NM_018335.4	ENSP00000399863	1
ZNF839	ENST00000561251	?	ENSP00000453137	1
ZNF839	ENST00000559185	NM_001267828.1	ENSP00000453109	1
ZNF839	ENST00000557803	?	?	1
ZNF839	ENST00000559155	?	ENSP00000453415	1
ZNF839	ENST00000558462	?	ENSP00000454159	1
ZNF839	ENST00000558490	?	ENSP00000452819	1
ZNF839	ENST00000420933	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CINP	ENST00000559504	?	ENSP00000453846	2
CINP	ENST00000559514	?	ENSP00000453839	2