Locus 108 (rs1190552)

SNiPAcard

Block annotations

Block info

genomic range	chr14:102,838,957-102,995,068
block size	156,112 bp
variant count	37 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.804 [-3.848 – 1.045]	gene(s) hit or close-by	ANKRD9, RNU6-244P, TECPR2
phastCons	μ = 0.035 [0 – 0.433]	eQTL gene(s)	ANKRD9, CINP, TECPR2, ZNF839
GERP++	μ = -0.650 [-4.97 – 1.33]	potentially regulated gene(s)	ANKRD9
CADD score	μ = 3.867 [0.244 – 18.09]	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	muscularis mucosae	1.64×10^{-12} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	lung	3.07×10^{-10} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	atrial appendage	4.94×10^{-7} (p-value)	GTEx Portal V6	8
ANKRD9	?	ENSG00000156381	transformed fibroblasts	3.75×10^{-25} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	transformed fibroblasts	1.39×10^{-13} (p-value)	GTEx Portal V6	35
TECPR2	?	ENSG00000196663	tibial artery	2.31×10^{-7} (p-value)	GTEx Portal V6	16
CINP	?	ENSG00000100865	tibial artery	6.65×10^{-9} (p-value)	GTEx Portal V6	34
CINP	?	ENSG00000100865	breast	8.44×10^{-10} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	blood	8.51×10^{-10} (p-value)	GTEx Portal V6	35
CINP	?	ENSG00000100865	thyroid	3.32×10^{-10} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	skeletal muscle	8.18×10^{-14} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	transverse colon	5.61×10^{-8} (p-value)	GTEx Portal V6	33
CINP	?	ENSG00000100865	EBV lymphocytes	1.82×10^{-8} (p-value)	GTEx Portal V6	11
CINP	?	ENSG00000100865	unexposed skin	7.47×10^{-9} (p-value)	GTEx Portal V6	34
CINP	?	ENSG00000100865	sun exposed skin	7.80×10^{-15} (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	liver	2.33×10^{-8} (p-value)	GTEx Portal V6	12

CINP	?	ENSG00000100865	liver	3.23×10-8 (p-value)	GTEx Portal V6	13
CINP	?	ENSG00000100865	aorta	3.94×10-9 (p-value)	GTEx Portal V6	11
CINP	?	ENSG00000100865	left ventricle	3.40×10-8 (p-value)	GTEx Portal V6	13
CINP	?	ENSG00000100865	subcutaneous adipocytes	2.21×10-14 (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	visceral adipocytes	1.70×10-12 (p-value)	GTEx Portal V6	36
CINP	?	ENSG00000100865	tibial nerve	4.97×10-10 (p-value)	GTEx Portal V6	35
CINP	?	ENSG00000100865	sigmoid colon	7.70×10-9 (p-value)	GTEx Portal V6	35
CINP	?	ENSG00000100865	esophagus mucosa	2.53×10-15 (p-value)	GTEx Portal V6	35
CINP	?	ENSG00000100865	gastroesophageal junction	7.26×10-8 (p-value)	GTEx Portal V6	17
CINP	?	ENSG00000100865	testis	8.62×10-8 (p-value)	GTEx Portal V6	12
ZNF839	ENST00000558490	ILMN_2186858	skin	1.56×10-7 (p-value)	MuTHER consortium	4
ZNF839	ENST00000420933		adipocyte	9.15×10-6 (p-value)	MuTHER consortium	4
ZNF839	ENST00000557803					
ZNF839	ENST00000559098					
ZNF839	ENST00000561251					
ZNF839	ENST00000558850					
ZNF839	ENST00000559155					
ZNF839	ENST00000559185					
ZNF839	ENST00000442396					
ZNF839	ENST00000560112					
ZNF839	ENST00000558462					
CINP	ENST00000216756	ILMN_1765257	blood	2.69×10-5 (p-value)	Westra et al.	3
CINP	ENST00000541568					
CINP	ENST00000559504					
CINP	ENST00000559514					
CINP	ENST00000536961					
ANKRD9	ENST00000559404	ILMN_1675937	blood	5.57×10-7 (p-value)	Westra et al.	2
ANKRD9	ENST00000286918					
ANKRD9	ENST00000559651					
ANKRD9	ENST00000557902					
ANKRD9	ENST00000560748					
CINP	?	ENSG00000100865	coronary artery	3.44×10-6 (p-value)	GTEx Portal V6	2
ZNF839	ENST00000420933	ILMN_2186858	b-cell	1.27×10-4 (p-value)	Fairfax et al.	1
ZNF839	ENST00000558490					
ZNF839	ENST00000557803					
ZNF839	ENST00000559098					
ZNF839	ENST00000559155					
ZNF839	ENST00000558850					
ZNF839	ENST00000561251					
ZNF839	ENST00000559185					
ZNF839	ENST00000442396					
ZNF839	ENST00000560112					
ZNF839	ENST00000558462					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000159684	1	ENST00000559404, ENST00000557902, ENST00000559651	ANKRD9

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
ENSR00001625365 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, CTCF, H2AZ, Rad21, Egr1
		skin (NHDF-AD)	H3K4me3, CTCF, DNase1, H3K4me2
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, DNase1
		blood (GM12878)	DNase1, H2AZ, Yy1, Rad21, CTCF
		lung (IMR90)	H3K4me2, H3K36me3, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF, H3K36me3
		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
ENSR00000181837 (CTCF binding site)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	DNase1, CTCF, H3K36me3, Rad21
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	CTCF, Max
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, H3K4me1, Rad21, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	CTCF, H3K36me3
ENSR00001460390 (promoter flanking region)	1	NHLF	H3K36me3, H3K27ac
		embryonic stem cell (H1ESC)	DNase1

		HSMMtube	H3K36me3
		Osteobl	H3K27ac, H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	Max
		skin (NHDF-AD)	H3K27ac, DNase1
		muscle (HSMM)	H2AZ, H3K27ac, H3K36me3, DNase1
		breast (HMEC)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	H3K27ac, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, Max, Cjun
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3, H3K9ac, H3K27ac, H3K4me1
ENSR00001625371 (promoter)	2	embryonic stem cell (H1ESC)	PolII, H3K9ac, CTCF, Yy1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me3, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, H3K4me2, PolII, H2AZ, H3K9ac, H3K27ac, Egr1
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me2, H3K4me1, PolII, FOXA1, H3K36me3, H3K4me3, H3K27ac, H3K9ac, DNase1
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, DNase1
		blood (GM12878)	H2AZ, H3K4me3, H3K4me2, DNase1
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, CTCF
		NHLF	DNase1
		Osteobl	H3K27ac, H3K4me3, H3K4me2, DNase1
		breast (HMEC)	H3K4me2, H3K4me3, DNase1
		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF, PolII, H3K4me3, H3K27ac, H3K4me2, H3K9ac, Ini1
		monocytes (Monocytes-CD14+)	H3K4me3, DNase1
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, H3K4me3
		A549	H3K4me3, H3K4me2, DNase1
ENSR00000181844 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, Egr1, CTCF, Yy1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	H3K4me3, CTCF, ELF1, H2AZ, H3K9ac, Rad21, ZBTB7A, Egr1
		skin (NHDF-AD)	CTCF, H3K4me2, H3K4me3
		muscle (HSMM)	H2AZ, CTCF, H3K4me3
		liver (HepG2)	DNase1, H3K4me3, CTCF, H3K4me2, Rad21
		lung (IMR90)	H3K4me2, H3K4me3, CTCF
		blood (GM12878)	DNase1, H2AZ, ELF1, Rad21, H3K4me3, H3K4me2, CTCF
		nervous (NH-A)	DNase1, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
		NHLF	H3K9ac, CTCF
		Osteobl	H3K4me3, H3K4me2, CTCF
		blood (DND-41)	H3K4me2, CTCF, H3K27me3
		breast (HMEC)	H3K4me2, H3K4me3, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K4me3, H3K4me2
		monocytes (Monocytes-CD14+)	CTCF, H3K4me2, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K4me3, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF
ENSR00000424062 (promoter flanking region)	1	embryonic stem cell (H1ESC)	SP1, USF1, H3K4me3, H3K4me2, TAF1, H3K27me3, PolII, Rad21, Gabp, TAF7, CTCF, Yy1, HDAC2, DNase1
		HSMMtube	CTCF, H2AZ, DNase1
		blood (K562)	Cfos, TAF1, H2AZ, HEY1, USF1, H3K9ac, Rad21, H3K27ac, PolII, H3K4me2, CTCF, H3K4me3, DNase1
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	DNase1, H3K4me3, CTCF, H2AZ
		liver (HepG2)	H3K4me2, USF1, TAF1, Rad21, PolII, H3K27ac, CTCF, H3K4me3, DNase1
		blood (GM12878)	EBF1, PolII, H2AZ, DNase1, Yy1, Rad21, H3K4me3, USF1, Cfos, CTCF
		lung (IMR90)	DNase1, H3K4me2, H4K5ac, H3K4me3, H3K9ac, CTCF
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	H3K27ac, H3K4me1, H3K9ac, H3K4me2, DNase1, CTCF, H3K4me3
		NHLF	H3K4me3, DNase1, CTCF, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	CTCF, H3K4me2, H3K4me3, H3K27ac, DNase1

cervix (HeLa-S3)	DNase1, CTCF, PolII, Cfos, Max, TAF1, H3K4me3, H3K27ac, H3K4me2, H3K9ac
monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K27ac, H3K4me3
endothelium (HUVEC)	H3K36me3, DNase1, CTCF, PolII, H3K4me2, H3K4me3, Max
A549	H3K9ac, CTCF, DNase1, H3K27ac, H3K4me2, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANKRD9	downstream gene variant, upstream gene variant	418	ENST00000559651	?	ENSP00000454100	4
ANKRD9	downstream gene variant, upstream gene variant	2632	ENST00000560748	?	ENSP00000453650	2
ANKRD9	downstream gene variant, upstream gene variant	2101	ENST00000286918	NM_152326.2	ENSP00000286918	6
ANKRD9	downstream gene variant, upstream gene variant	324	ENST00000557902	?	ENSP00000454080	4
ANKRD9	downstream gene variant, upstream gene variant	83	ENST00000559404	?	ENSP00000453417	3
RNU6-244P	upstream gene variant, downstream gene variant	2017	ENST00000391028	?	?	6
TECPR2	downstream gene variant	1663	ENST00000561228	?	?	1
TECPR2	upstream gene variant, downstream gene variant	236	ENST00000559124	?	?	6
TECPR2	upstream gene variant	435	ENST00000557786	?	?	1
TECPR2	upstream gene variant, downstream gene variant	486	ENST00000561099	?	?	5
TECPR2	downstream gene variant	1339	ENST00000558678	NM_001172631.1	ENSP00000453671	1
TECPR2	downstream gene variant	1763	ENST00000359520	NM_014844.3	ENSP00000352510	1
TECPR2	upstream gene variant, downstream gene variant	1211	ENST00000560060	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANKRD9	ENST00000560748	?	ENSP00000453650	2
ANKRD9	ENST00000286918	NM_152326.2	ENSP00000286918	2
TECPR2	ENST00000558678	NM_001172631.1	ENSP00000453671	20
TECPR2	ENST00000359520	NM_014844.3	ENSP00000352510	25
TECPR2	ENST00000561099	?	?	1
TECPR2	ENST00000561228	?	?	11
TECPR2	ENST00000560060	?	?	3
TECPR2	ENST00000557786	?	?	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANKRD9	ENST00000286918	NM_152326.2	ENSP00000286918	1
TECPR2	ENST00000359520	NM_014844.3	ENSP00000352510	4

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANKRD9	ENST00000559404	?	ENSP00000453417	1