Locus 102 (rs4951168)

SNiPAcard

Block annotations

Block info

genomic range	chr1:205,014,930-205,235,781
block size	220,852 bp
variant count	55 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.887 [-3.535 – 2.059]	**gene(s) hit or close-by**	CNTN2, DSTYK, RBBP5, RP11-383G10.1, RP11-383G10.5, RP11-536L3.4, TMCC2, TMEM81
phastCons	μ = 0.040 [0 – 1]	**eQTL gene(s)**	CNTN2, DSTYK, RBBP5, RP11-383G10.5, TMEM81
GERP++	μ = -0.627 [-9.79 – 5.76]	**potentially regulated gene(s)**	CNTN2, DSTYK, RBBP5
CADD score	μ = 3.523 [0.073 – 22.1]	**disease gene(s)**	CNTN2, DSTYK

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Body weight	9.92×10^{-5}	dbGaP	pha003026	1
Waist circumference	7.71×10^{-5}	dbGaP	pha003024	4
Stroke	2.00×10^{-4}	dbGaP	pha002886	2

Disease gene annotation

gene	trait	source DB	source entry/link
CNTN2	EPILEPSY, FAMILIAL ADULT MYOCLONIC, 5	OMIM	MIM:615400
DSTYK	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT, SUSCEPTIBILITY [...]	OMIM	MIM:610805
CNTN2	Benign adult familial myoclonic epilepsy	OrphaNet	OrphaNet:86814
DSTYK	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT, CAKUT1	DECIPHER	MIM:610805

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)
TMEM81	missense variant	ENST00000367167	NM_203376.1	ENSP00000356135	R/Q	cGg/cAg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CNTN2	?	ENSG00000184144	thyroid	2.07×10^{-8} (p-value)	GTEx Portal V6	36
TMEM81	?	ENSG00000174529	skeletal muscle	9.35×10^{-8} (p-value)	GTEx Portal V6	34
RBBP5	?	ENSG00000117222	tibial nerve	1.19×10^{-5} (p-value)	GTEx Portal V6	4
CNTN2	?	ENSG00000184144	tibial nerve	1.17×10^{-6} (p-value)	GTEx Portal V6	4

CNTN2	?	ENSG00000184144	muscularis mucosae	3.47×10-7 (p-value)	GTEx Portal V6	43
TMEM81	?	ENSG00000174529	muscularis mucosae	1.83×10-5 (p-value)	GTEx Portal V6	2
RP11-383G10.5	?	ENSG00000225063	testis	6.32×10-9 (p-value)	GTEx Portal V6	32
CNTN2	?	ENSG00000184144	sun exposed skin	1.96×10-6 (p-value)	GTEx Portal V6	8
DSTYK	ENST00000367162	ILMN_2352023	monocyte	1.04×10-30 (p-value)	Zeller et al.	5
DSTYK	ENST00000367161					
CNTN2	ENST00000331830	ILMN_1718896	b-cell	3.40×10-4 (p-value)	Fairfax et al.	4

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000052358	2	ENCP00000006264	RBBP5
		ENCP00000006267	DSTYK
		ENCP00000006260	CNTN2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000082308 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Max, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	H2AZ, Rad21, CTCF
		lung (IMR90)	CTCF, DNase1
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	DNase1, CTCF, H3K4me3
ENSR00001589901 (promoter flanking region)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Max, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	H2AZ, Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K4me3, CTCF, DNase1
ENSR00000288565	1	embryonic stem cell (H1ESC)	H3K36me3

(open chromatin region)		lung (IMR90)	H3K36me3
		blood (K562)	DNase1
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000551432 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, CTCF, DNase1
		HSMMtube	H3K27ac, DNase1
		blood (K562)	DNase1, FOSL1, Cjun, H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K27ac
		muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3, DNase1
		lung (IMR90)	DNase1, H3K27ac, H3K36me3
		nervous (NH-A)	DNase1, H3K27ac, H3K9ac
		skin (NHEK)	H3K4me1, H3K27ac, DNase1
		NHLF	H3K27ac, H3K4me1, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Max, Cmyc, Cfos, H3K36me3, H3K27ac, Jund, Cjun, H3K9ac
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac, H3K36me3
ENSR00001527878 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001589908 (CTCF binding site)	1	NHLF	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, CTCF
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001527883 (enhancer)	1	cervix (HeLa-S3)	H3K27me3
		HSMMtube	H3K27ac, DNase1
		liver (HepG2)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00000288582 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1, Jund
		blood (K562)	DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CNTN2	downstream gene variant	270	ENST00000481872	?	?	4
CNTN2	upstream gene variant, downstream gene variant	328	ENST00000525433	?	?	7
CNTN2	downstream gene variant	1409	ENST00000530594	?	?	2
CNTN2	downstream gene variant	756	ENST00000331830	NM_005076.3	ENSP00000330633	2
CNTN2	downstream gene variant	2970	ENST00000527340	?	?	1
DSTYK	downstream gene variant, upstream gene variant	704	ENST00000367162	NM_015375.2	ENSP00000356130	4
DSTYK	downstream gene variant, upstream gene variant	704	ENST00000367161	NM_199462.2	ENSP00000356129	4
DSTYK	downstream gene variant, upstream gene variant	849	ENST00000615388	?	ENSP00000478016	3
RBBP5	downstream gene variant, upstream gene variant	108	ENST00000367164	NM_001193272.1	ENSP00000356132	7

RBBP5	downstream gene variant, upstream gene variant	197	ENST00000484379	?	?	4
RBBP5	downstream gene variant, upstream gene variant	108	ENST00000264515	NM_005057.3, NM_001193273.1	ENSP00000264515	7
RP11-383G10.1	upstream gene variant, downstream gene variant	312	ENST00000428506	?	?	6
RP11-383G10.5	downstream gene variant, upstream gene variant	3749	ENST00000452599	?	?	2
RP11-536L3.4	upstream gene variant, downstream gene variant	540	ENST00000603227	?	?	5
TMCC2	upstream gene variant	3480	ENST00000330675	NM_001297611.1	ENSP00000331842	1
TMCC2	upstream gene variant	3356	ENST00000367159	?	ENSP00000356127	1
TMEM81	downstream gene variant, upstream gene variant	1482	ENST00000367167	NM_203376.1	ENSP00000356135	5

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TMEM81	ENST00000367167	NM_203376.1	ENSP00000356135	L	ctC/ctT	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN2	ENST00000331830	NM_005076.3	ENSP00000330633	5
CNTN2	ENST00000481872	?	?	4
DSTYK	ENST00000615388	?	ENSP00000478016	3
DSTYK	ENST00000367161	NM_199462.2	ENSP00000356129	14
DSTYK	ENST00000367162	NM_015375.2	ENSP00000356130	14
RBBP5	ENST00000484379	?	?	4
RBBP5	ENST00000367164	NM_001193272.1	ENSP00000356132	8
RBBP5	ENST00000264515	NM_005057.3, NM_001193273.1	ENSP00000264515	8
RP11-383G10.5	ENST00000452599	?	?	1
TMCC2	ENST00000495538	?	?	6
TMCC2	ENST00000358024	NM_014858.3	ENSP00000350718	6
TMCC2	ENST00000367159	?	ENSP00000356127	2
TMCC2	ENST00000330675	NM_001297611.1	ENSP00000331842	2
TMCC2	ENST00000545499	NM_001242925.1	ENSP00000437943	6
TMCC2	ENST00000329800	NM_001297613.1	ENSP00000329436	1
TMCC2	ENST00000468846	?	?	1
TMCC2	ENST00000481950	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN2	ENST00000331830	NM_005076.3	ENSP00000330633	3
RBBP5	ENST00000367164	NM_001193272.1	ENSP00000356132	2
RBBP5	ENST00000264515	NM_005057.3, NM_001193273.1	ENSP00000264515	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TMCC2	ENST00000495538	?	1