Locus 432 (rs41264869)

SNiPAcard

Block annotations

Block info

genomic range	chr1:205,028,581-205,087,683
block size	59,103 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.211 [-1.802 – 1.411]	gene(s) hit or close-by	AL583832.1, CNTN2, RBBP5, RP11-536L3.4, TMEM81
phastCons	μ = 0.014 [0 – 0.121]	eQTL gene(s)	CNTN2, DSTYK, RBBP5, TMCC2
GERP++	μ = -0.422 [-5.23 – 3.62]	potentially regulated gene(s)	CNTN2
CADD score	μ = 3.701 [0.3 – 8.736]	disease gene(s)	CNTN2, DSTYK

Trait annotations

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 regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RBBP5	?	ENSG00000117222	transformed fibroblasts	7.58×10^{-6} (p-value)	GTEx Portal V6	1
RBBP5	?	ENSG00000117222	tibial artery	4.05×10^{-6} (p-value)	GTEx Portal V6	1
TMCC2	?	ENSG00000133069	tibial artery	1.25×10^{-5} (p-value)	GTEx Portal V6	2
RBBP5	?	ENSG00000117222	thyroid	7.76×10^{-6} (p-value)	GTEx Portal V6	12
DSTYK	ENST00000367162	ILMN_2352023	monocyte	1.18×10^{-32} (p-value)	Zeller et al.	1
DSTYK	ENST00000367161					
CNTN2	?	ENSG00000184144	unexposed skin	9.06×10^{-6} (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
RBBP5	ENST00000264515	205169_at	chr1	blood	7.50×10^{-8} (p-value)	Dixon et al.	1
RBBP5	ENST00000367164						

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000006260		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000167977 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF, DNase1, H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3, DNase1
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL583832.1	upstream gene variant, downstream gene variant	274	ENST00000515887	?	?	4
CNTN2	downstream gene variant	855	ENST00000331830	NM_005076.3	ENSP00000330633	2
CNTN2	upstream gene variant, downstream gene variant	486	ENST00000525433	?	?	5
CNTN2	downstream gene variant	1039	ENST00000532366	?	?	4
CNTN2	upstream gene variant, downstream gene variant	100	ENST00000527340	?	?	3
CNTN2	upstream gene variant, downstream gene variant	1937	ENST00000530594	?	?	5
CNTN2	downstream gene variant	758	ENST00000481872	?	?	4
CNTN2	downstream gene variant	281	ENST00000530117	?	?	2
RP11-536L3.4	downstream gene variant	460	ENST00000603227	?	?	2
TMEM81	downstream gene variant	3641	ENST00000367167	NM_203376.1	ENSP00000356135	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN2	ENST00000481872	?	?	8
CNTN2	ENST00000525433	?	?	1
CNTN2	ENST00000530117	?	?	2
CNTN2	ENST00000331830	NM_005076.3	ENSP00000330633	8
CNTN2	ENST00000527340	?	?	2
RBBP5	ENST00000264515	NM_005057.3, NM_001193273.1	ENSP00000264515	7
RBBP5	ENST00000367164	NM_001193272.1	ENSP00000356132	7
RBBP5	ENST00000484379	?	?	5

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNTN2	ENST00000331830	NM_005076.3	ENSP00000330633	2

CNTNAP2	ENST00000538538	NM_014141.6	ENSP00000438699	2

Locus 432 (sentinel SNP rs41264869 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.