Locus 445 (rs4453827)

SNiPAcard

Block annotations

Block info

genomic range	chr3:53,741,287-53,789,698
block size	48,412 bp
variant count	25 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.528 [-3.374 – 1.263]	gene(s) hit or close-by	CACNA1D
phastCons	μ = 0.022 [0 – 0.396]	eQTL gene(s)	CHDH, IL17RB
GERP++	μ = -1.717 [-8.88 – 4.79]	potentially regulated gene(s)	DCP1A
CADD score	μ = 3.756 [0.136 – 16.38]	disease gene(s)	CACNA1D

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CACNA1D	Primary aldosteronism, seizures, and neurologic abnormalities	DECIPHER	MIM:615474
CACNA1D	Sinoatrial node dysfunction and deafness	DECIPHER	MIM:614896
CACNA1D	SINOATRIAL NODE DYSFUNCTION AND DEAFNESS	OMIM	MIM:614896
CACNA1D	PRIMARY ALDOSTERONISM, SEIZURES, AND NEUROLOGIC [...]	OMIM	MIM:615474
CACNA1D	Sinoatrial node dysfunction and deafness	OrphaNet	OrphaNet:324321
CACNA1D	Aldosterone-producing adenoma with seizures and neurological abnormalities	OrphaNet	OrphaNet:369929

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CHDH	?	ENSG00000016391	tibial nerve	4.41×10^{-8} (p-value)	GTEx Portal V6	24
IL17RB	?	ENSG00000056736	tibial nerve	3.84×10^{-9} (p-value)	GTEx Portal V6	24

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000340690	1	ENCP00000036821	DCP1A

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001479254 (enhancer)	3	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001364519 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3
		blood (GM12878)	H3K4me1, H3K27ac, H3K4me2
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CACNA1D	downstream gene variant	2904	ENST00000481085	?	ENSP00000418045	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CACNA1D	ENST00000350061	NM_001128840.2	ENSP00000288133	25
CACNA1D	ENST00000288139	NM_000720.3	ENSP00000288139	25
CACNA1D	ENST00000481085	?	ENSP00000418045	16
CACNA1D	ENST00000422281	NM_001128839.2	ENSP00000409174	25
CACNA1D	ENST00000481478	?	ENSP00000418014	25