

SNiPACard

Block annotations

Block info







genomic range	chr3:53,741,287-53,789,698 <i>e!</i>
block size	48,412 bp
variant count	25 variants

Basic features

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



Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CACNA1D <i>e!</i>	Primary aldosteronism, seizures, and neurologic abnormalities	DECIPHER	MIM:615474 
CACNA1D <i>e!</i>	Sinoatrial node dysfunction and deafness	DECIPHER	MIM:614896 
CACNA1D <i>e!</i>	SINOATRIAL NODE DYSFUNCTION AND DEAFNESS	OMIM	MIM:614896 
CACNA1D <i>e!</i>	PRIMARY ALDOSTERONISM, SEIZURES, AND NEUROLOGIC [...]	OMIM	MIM:615474 
CACNA1D <i>e!</i>	Sinoatrial node dysfunction and deafness	OrphaNet	OrphaNet:324321 
CACNA1D <i>e!</i>	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 <i>e!</i>	?	ENSG00000016391 <i>e!</i>	tibial nerve	4.41×10 ⁻⁸ (p-value)	GTEx Portal V6 	24
IL17RB <i>e!</i>	?	ENSG00000056736 <i>e!</i>	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)
ENCE000000340690 <i>e!</i>	1	ENCP00000036821	DCP1A <i>e!</i>

Regulatory feature cluster

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

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CACNA1D <i>e!</i>	downstream gene variant	2904	ENST00000481085 <i>e!</i>	?	ENSP00000418045 <i>e!</i>	2

Putative effect on transcript

Intron variant

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

