SNiPAcard

Block annotations

Block info

genomic range	chr11:12,038,874-12,050,652
block size	11,779 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.783 [0.562 – 1.004]	**gene(s) hit or close-by**	RP13-631K18.2
phastCons	μ = 0.040 [0.009 – 0.072]	**eQTL gene(s)**	DKK3, USP47
GERP++	μ = 1.406 [-0.558 – 3.37]	**potentially regulated gene(s)**	MICALCL, RP11-483L5.1
CADD score	μ = 4.825 [2.084 – 7.565]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DKK3	?	ENSG00000050165	gastroesophageal junction	2.41×10^{-6} (p-value)	GTEx Portal V6	2
DKK3	?	ENSG00000050165	muscularis mucosae	4.15×10^{-13} (p-value)	GTEx Portal V6	2
DKK3	?	ENSG00000050165	skeletal muscle	3.01×10^{-6} (p-value)	GTEx Portal V6	1
DKK3	?	ENSG00000050165	tibial artery	1.80×10^{-32} (p-value)	GTEx Portal V6	2
USP47	?	ENSG00000170242	tibial artery	4.65×10^{-6} (p-value)	GTEx Portal V6	1
DKK3	?	ENSG00000050165	aorta	1.52×10^{-23} (p-value)	GTEx Portal V6	2
USP47	?	ENSG00000170242	aorta	3.77×10^{-16} (p-value)	GTEx Portal V6	2
DKK3	?	ENSG00000050165	thyroid	6.76×10^{-11} (p-value)	GTEx Portal V6	2
DKK3	?	ENSG00000050165	subcutaneous adipocytes	1.77×10^{-7} (p-value)	GTEx Portal V6	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000098145	1	ENCP00000011204	MICALCL
		ENCP00000011165	RP11-483L5.1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000317680 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	Rad21
		Osteobl	H3K4me2
		blood (K562)	H3K27me3, DNase1, MEF2A
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3

		endothelium (HUVEC)	DNase1, Cjun
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00000317681 e! (open chromatin region)	1	blood (K562)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP13-631K18.2 e!	upstream gene variant	1770	ENST00000531559 e!	?	?	1