Locus 129 (rs484915)

SNiPAcard

Block annotations

Block info

genomic range	chr11:102,671,697-102,673,248
block size	1,552 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.143 [-0.238 – -0.048]	gene(s) hit or close-by	AP000619.6, MMP1, WTAPP1
phastCons	μ = 0.001 [0.001 – 0.001]	eQTL gene(s)	MMP1, WTAPP1
GERP++	μ = -1.033 [-1.22 – -0.846]	potentially regulated gene(s)	DYNC2H1, MMP27, MMP3, RP11-817J15.3
CADD score	μ = 2.032 [1.406 – 2.658]	disease gene(s)	MMP3, DYNC2H1, MMP1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
MMP3	CORONARY HEART DISEASE, SUSCEPTIBILITY TO, 6	OMIM	MIM:614466
DYNC2H1	SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY	OMIM	MIM:613091
MMP1	Severe generalized recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79408
DYNC2H1	Short rib-polydactyly syndrome, Verma-Naumoff type	OrphaNet	OrphaNet:93271
DYNC2H1	Short rib-polydactyly syndrome, Majewski type	OrphaNet	OrphaNet:93269
DYNC2H1	Jeune syndrome	OrphaNet	OrphaNet:474
DYNC2H1	short rib-polydactyly syndrome type 3 (SRPS3)	DECIPHER	MIM:263510
DYNC2H1	asphyxiating thoracic dystrophy type 3 (ATD3)	DECIPHER	MIM:613091

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MMP1	?	ENSG00000196611	tibial nerve	1.40×10^{-5} (p-value)	GTEx Portal V6	2
MMP1	?	ENSG00000196611	transformed fibroblasts	1.64×10^{-13} (p-value)	GTEx Portal V6	2
WTAPP1	?	ENSG00000255282	transformed fibroblasts	2.04×10^{-11} (p-value)	GTEx Portal V6	2
MMP1	?	ENSG00000196611	left ventricle	2.57×10^{-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)
ENCE00000120249	1	ENCP00000014388	MMP27
		ENCP00000014401	MMP3
ENCE00000120235	1	ENCP00000014388	MMP27
		ENCP00000014387	RP11-817J15.3
		ENCP00000014401	MMP3
		ENCP00000014410	DYNC2H1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001572918 (promoter flanking region)	1	endothelium (HUVEC)	Max
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AP000619.6	downstream gene variant	4431	ENST00000546014	?	?	1
MMP1	upstream gene variant	2806	ENST00000315274	NM_002421.3, NM_001145938.1	ENSP00000322788	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
WTAPP1	ENST00000525739	?	?	2
WTAPP1	ENST00000371455	?	?	2
WTAPP1	ENST00000544704	?	?	2