Locus 140 (rs12052479)

SNiPAcard

Block annotations

Block info

genomic range	chr2:173,373,190-173,450,756
block size	77,567 bp
variant count	18 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.005 [-4.066 – 0.826]	gene(s) hit or close-by	AC093818.1, ITGA6, PDK1
phastCons	μ = 0.125 [0 – 0.539]	eQTL gene(s)	PDK1
GERP++	μ = -0.023 [-2.38 – 2]	potentially regulated gene(s)	METAP1D, PDK1
CADD score	μ = 3.881 [0.345 – 10.73]	disease gene(s)	ITGA6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ITGA6	Epidermolysis Bullosa with Pyloric Atresia	DECIPHER	MIM:226730
ITGA6	EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA	OMIM	MIM:226730
ITGA6	Junctional epidermolysis bullosa - pyloric atresia	OrphaNet	OrphaNet:79403

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDK1	?	ENSG00000152256	skeletal muscle	2.06×10^{-11} (p-value)	GTEx Portal V6	15
PDK1	?	ENSG00000152256	blood	6.90×10^{-7} (p-value)	GTEx Portal V6	12
PDK1	?	ENSG00000152256	atrial appendage	1.89×10^{-6} (p-value)	GTEx Portal V6	3
PDK1	?	ENSG00000152256	left ventricle	4.16×10^{-7} (p-value)	GTEx Portal V6	3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000269288	1	ENCP00000028626	PDK1
		ENCP00000028606	METAP1D

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001658967 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PDK1	1	ENST00000282077	hsa-miR-33a-5p
		ENST00000392571	hsa-miR-33b-5p

ENST00000410055
ENST00000416991
ENST00000431718

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC093818.1	downstream gene variant	4197	ENST00000444919	?	?	2
AC093818.1	downstream gene variant, upstream gene variant	567	ENST00000416080	?	?	2
ITGA6	downstream gene variant	4134	ENST00000416789	?	ENSP00000388435	1
ITGA6	downstream gene variant	4210	ENST00000458358	?	ENSP00000394169	1
ITGA6	downstream gene variant	2232	ENST00000264107	NM_000210.2	ENSP00000264107	1
ITGA6	downstream gene variant	3928	ENST00000409532	?	ENSP00000386614	1
ITGA6	downstream gene variant	2180	ENST00000409080	NM_001079818.1	ENSP00000386896	1
ITGA6	downstream gene variant	4210	ENST00000442250	?	ENSP00000406694	1
PDK1	upstream gene variant	2127	ENST00000466437	?	?	1
PDK1	downstream gene variant	3771	ENST00000436490	?	ENSP00000406608	1
PDK1	downstream gene variant	3385	ENST00000443353	?	ENSP00000399558	1
PDK1	downstream gene variant	3385	ENST00000439519	?	ENSP00000388366	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC093818.1	ENST00000450443	?	?	15
AC093818.1	ENST00000442417	?	?	15
PDK1	ENST00000416991	?	ENSP00000399160	1
PDK1	ENST00000282077	?	ENSP00000282077	3
PDK1	ENST00000392571	NM_001278549.1	ENSP00000376352	3
PDK1	ENST00000410055	NM_002610.4	ENSP00000386985	3
PDK1	ENST00000431718	?	ENSP00000413629	1
PDK1	ENST00000466437	?	?	2