Locus 224 (rs922182)

SNiPAcard

Block annotations

Block info

genomic range	chr15:64,275,409-64,284,214
block size	8,806 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.239 [-1.896 – 0.659]	gene(s) hit or close-by	DAPK2
phastCons	μ = 0.041 [0 – 0.404]	eQTL gene(s)	DAPK2, RP11-111E14.1
GERP++	μ = -1.744 [-7.76 – 1.57]	potentially regulated gene(s)	–
CADD score	μ = 6.194 [0.564 – 12.3]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DAPK2	?	ENSG00000035664	aorta	2.07×10^{-13} (p-value)	GTEx Portal V6	9
RP11-111E14.1	?	ENSG00000259351	aorta	2.21×10^{-6} (p-value)	GTEx Portal V6	4
DAPK2	?	ENSG00000035664	muscularis mucosae	4.12×10^{-6} (p-value)	GTEx Portal V6	4
DAPK2	?	ENSG00000035664	tibial artery	2.45×10^{-5} (p-value)	GTEx Portal V6	1
RP11-111E14.1	?	ENSG00000259351	tibial nerve	2.91×10^{-5} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
CTCF	MA0139.1	15	no	0.000	1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000021558 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF
		blood (GM12878)	Rad21, CTCF, H3K36me3
		blood (K562)	Rad21, CTCF
		skin (NHEK)	CTCF
ENSR00000409563 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	DNase1, Rad21, BCL11A, H3K27ac, H3K4me2, H3K4me1, BATF, PU1, PolII
		blood (K562)	DNase1
		skin (NHDF-AD)	DNase1
ENSR00001023121 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	H3K4me1, H3K27ac
		lung (IMR90)	H3K27me3
		blood (K562)	PU1
ENSR00001453015 (promoter flanking region)	1	blood (GM12878)	H3K4me1, H3K27ac
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DAPK2	downstream gene variant	159	ENST00000559306	?	ENSP00000452970	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DAPK2	ENST00000561162	?	ENSP00000453284	11
DAPK2	ENST00000559897	?	?	11
DAPK2	ENST00000261891	?	ENSP00000261891	11
DAPK2	ENST00000559007	?	ENSP00000453639	11
DAPK2	ENST00000457488	NM_014326.3	ENSP00000408277	11
DAPK2	ENST00000558069	?	ENSP00000453235	11
DAPK2	ENST00000558482	?	?	11
DAPK2	ENST00000612884	?	ENSP00000484390	11
DAPK2	ENST00000559306	?	ENSP00000452970	9