Locus 435 (rs530022)

SNiPAcard

Block annotations

Block info

genomic range	chr1:168,664,450-168,675,971
block size	11,522 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.033 [-0.449 – 0.632]	gene(s) hit or close-by	DPT
phastCons	μ = 0.002 [0 – 0.007]	eQTL gene(s)	DPT
GERP++	μ = -0.783 [-2.66 – 1.57]	potentially regulated gene(s)	RP5-1018K9.1, TBX19
CADD score	μ = 2.879 [1.17 – 5.142]	disease gene(s)	TBX19

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TBX19	ACTH DEFICIENCY, ISOLATED	OMIM	MIM:201400
TBX19	Congenital isolated ACTH deficiency	OrphaNet	OrphaNet:199296

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DPT	ENST00000367817	ILMN_1708107	skin	1.29×10-6 (p-value)	MuTHER consortium	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000043270	1	ENCP00000005412	TBX19
		ENCP00000005426	RP5-1018K9.1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DPT	downstream gene variant	247	ENST00000367817	NM_001937.4	ENSP00000356791	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DPT	ENST00000367817	NM_001937.4	ENSP00000356791	9