Locus 297 (rs494189)

SNiPAcard

Block annotations

Block info

genomic range	chr1:168,696,549-168,729,840
block size	33,292 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.151 [-4.415 – 0.9]	gene(s) hit or close-by	AL049798.1, DPT, RP1-10C16.1
phastCons	μ = 0.007 [0 – 0.091]	eQTL gene(s)	DPT
GERP++	μ = -1.886 [-6.87 – 0.0691]	potentially regulated gene(s)	DPT
CADD score	μ = 1.904 [0.114 – 6.621]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DPT	ENST00000367817	ILMN_1708107	skin	1.26×10^{-5} (p-value)	MuTHER consortium	3
DPT	?	ENSG00000143196	aorta	1.19×10^{-6} (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000043396	1	ENCP00000005421	DPT

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001587687 (enhancer)	1	HSMMtube	H3K27me3
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me3, H3K4me2
ENSR00000547238 (TF binding site)	1	cervix (HeLa-S3)	DNase1
		embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	H3K36me3
		A549	H3K4me3, H3K4me2
		nervous (NH-A)	DNase1
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL049798.1	downstream gene variant, upstream gene variant	2456	ENST00000408651	?	?	4
DPT	upstream gene variant	1632	ENST00000367817	NM_001937.4	ENSP00000356791	2
RP1-10C16.1	downstream gene variant	2763	ENST00000457405	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DPT	ENST00000367817	NM_001937.4	ENSP00000356791	3