

SNiPAcad

Block annotations

Block info





genomic range	chr17:26,610,513-26,610,513 <i>e!</i>
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.216	gene(s) hit or close-by	KRT18P55 <i>e!</i>
phastCons	0.996	eQTL gene(s)	SARM1 <i>e!</i> , TMEM199 <i>e!</i> , TMEM97 <i>e!</i>
GERP++	-2.77	potentially regulated gene(s)	-
CADD score	1.06	disease gene(s)	-

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SARM1 <i>e!</i>	ENST00000585482 <i>e!</i>	ILMN_1746265 <i>e!</i>	adipocyte	1.70×10 ⁻⁶ (p-value)	MuTHER consortium 	1
TMEM97 <i>e!</i>	ENST00000226230 <i>e!</i>	ILMN_1710962 <i>e!</i>	blood	1.33×10 ⁻⁵ (p-value)	MuTHER consortium 	1
TMEM97 <i>e!</i>	ENST00000336687 <i>e!</i>		adipocyte	4.53×10 ⁻⁷ (p-value)	MuTHER consortium 	1
TMEM97 <i>e!</i>	ENST00000582384 <i>e!</i>					
TMEM199 <i>e!</i>	ENST00000483505 <i>e!</i>	ILMN_1748481 <i>e!</i>	blood	2.43×10 ⁻⁵ (p-value)	Westra et al. 	1
TMEM199 <i>e!</i>	ENST00000509083 <i>e!</i>					
TMEM199 <i>e!</i>	ENST00000585027 <i>e!</i>					
TMEM199 <i>e!</i>	ENST00000292114 <i>e!</i>					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001535058 <i>e!</i> (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
KRT18P55 <i>e!</i>	ENST00000577198 <i>e!</i>	?	?	1
KRT18P55 <i>e!</i>	ENST00000578578 <i>e!</i>	?	?	1

