Locus 415 (rs4851016)

SNiPAcard

Block annotations

Block info

genomic range	chr2:103,098,191-103,120,889
block size	22,699 bp
variant count	56 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.463 [-5.522 – 2.646]	gene(s) hit or close-by	SLC9A4
phastCons	μ = 0.013 [0 – 0.34]	eQTL gene(s)	AC007278.2, AC007278.3, IL18R1, IL18RAP
GERP++	μ = -0.487 [-4.99 – 3.15]	potentially regulated gene(s)	SLC9A4, TMEM182
CADD score	μ = 2.505 [0.007 – 12.24]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC007278.3	?	ENSG00000234389	lung	5.32×10^{-7} (p-value)	GTEx Portal V6	54
IL18R1	?	ENSG00000115604	sun exposed skin	6.92×10^{-6} (p-value)	GTEx Portal V6	18
AC007278.2	?	ENSG00000236525	blood	1.65×10^{-8} (p-value)	GTEx Portal V6	54
IL18RAP	?	ENSG00000115607	blood	1.07×10^{-14} (p-value)	GTEx Portal V6	54
AC007278.3	?	ENSG00000234389	blood	3.12×10^{-12} (p-value)	GTEx Portal V6	54
AC007278.3	?	ENSG00000234389	subcutaneous adipocytes	4.21×10^{-6} (p-value)	GTEx Portal V6	33

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000027326		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000257544	2	ENCP00000027332	TMEM182

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001544967 (promoter flanking region)	3	NHLF	DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3

	nervous (NH-A)	DNase1
	A549	H3K27me3
	skin (NHEK)	DNase1, H3K4me2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SLC9A4	upstream gene variant	872	ENST00000492154	?	?	28

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	56
SLC9A4	ENST00000492154	?	?	7