Locus 168 (rs6543140)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,991,163-103,092,575
block size	101,413 bp
variant count	43 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.097 [-2.8 – 1.633]	gene(s) hit or close-by	AC007248.7, AC007278.2, AC007278.3, IL18R1, IL18RAP, MIR4772, SLC9A4
phastCons	μ = 0.055 [0 – 1]	eQTL gene(s)	AC007278.3, IL18R1, IL18RAP, IL1RL1
GERP++	μ = -0.748 [-6.53 – 3.67]	potentially regulated gene(s)	IL1R2, SLC9A2, SLC9A4
CADD score	μ = 3.166 [0.068 – 20.9]	disease gene(s)	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AC007278.3	?	ENSG00000234389	visceral adipocytes	5.97×10^{-7} (p-value)	GTEx Portal V6	30
AC007278.3	?	ENSG00000234389	lung	3.09×10^{-12} (p-value)	GTEx Portal V6	42
IL1RL1	?	ENSG00000115602	lung	9.84×10^{-8} (p-value)	GTEx Portal V6	41
AC007278.3	?	ENSG00000234389	tibial nerve	6.35×10^{-7} (p-value)	GTEx Portal V6	39
AC007278.3	?	ENSG00000234389	atrial appendage	2.87×10^{-6} (p-value)	GTEx Portal V6	13
IL18RAP	?	ENSG00000115607	blood	1.78×10^{-9} (p-value)	GTEx Portal V6	42
AC007278.3	?	ENSG00000234389	blood	9.14×10^{-20} (p-value)	GTEx Portal V6	42
IL18R1	?	ENSG00000115604	thyroid	1.71×10^{-5} (p-value)	GTEx Portal V6	9
AC007278.3	?	ENSG00000234389	thyroid	6.60×10^{-7} (p-value)	GTEx Portal V6	42
AC007278.3	?	ENSG00000234389	esophagus mucosa	4.79×10^{-7} (p-value)	GTEx Portal V6	42
AC007278.3	?	ENSG00000234389	subcutaneous adipocytes	7.55×10^{-6} (p-value)	GTEx Portal V6	13

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	8	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000027325		

ENCODE promoter-associated distal DHS (Enhancer)

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

ENCE00000257249	2	ENCP00000027299	IL1R2
		ENCP00000027329	SLC9A2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001655327 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		HSMMtube	H3K27me3
		A549	H3K27me3
		blood (K562)	H2AZ
ENSR00001544959 (promoter flanking region)	1	NHLF	DNase1
		liver (HepG2)	H3K27me3
		HSMMtube	H3K27me3, DNase1
		blood (DND-41)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	DNase1
		muscle (HSMM)	DNase1
ENSR00000595671 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3
ENSR00000677146 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	CTCF
		A549	H3K27me3
ENSR00001544961 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3, CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF, H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, H3K27me3
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		A549	CTCF, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC007248.7	downstream gene variant	383	ENST00000430344	?	?	3
AC007278.2	upstream gene variant, downstream gene variant	1679	ENST00000436582	?	?	2
AC007278.3	downstream gene variant	733	ENST00000450893	?	?	1

IL18R1	downstream gene variant	722	ENST00000334376	?	ENSP00000334030	1
IL18R1	downstream gene variant	4746	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	1
IL18R1	downstream gene variant	4763	ENST00000409599	?	ENSP00000387211	1
IL18RAP	upstream gene variant	1942	ENST00000450855	?	ENSP00000389815	2
IL18RAP	upstream gene variant, downstream gene variant	1150	ENST00000264260	NM_003853.2	ENSP00000264260	8
IL18RAP	upstream gene variant, downstream gene variant	1331	ENST00000409369	?	ENSP00000387201	7
MIR4772	upstream gene variant	11	ENST00000581495	?	?	1
SLC9A4	upstream gene variant	189	ENST00000295269	NM_001011552.3	ENSP00000295269	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL18R1	ENST00000409599	?	ENSP00000387211	2
IL18R1	ENST00000410040	?	ENSP00000386663	2
IL18R1	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	2
IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	9
IL18RAP	ENST00000409369	?	ENSP00000387201	9
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC007278.3	ENST00000450893	?	1