Locus 19 (rs2058622)

SNiPAcard

Block annotations

Block info

genomic range	chr2:102,918,018-103,093,243
block size	175,226 bp
variant count	94 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.815 [-4.683 – 1.369]	gene(s) hit or close-by	AC007248.7, AC007278.2, AC007278.3, IL18R1, IL18RAP, IL1RL1, MIR4772, SLC9A4
phastCons	μ = 0.030 [0 – 0.992]	eQTL gene(s)	AC007248.7, AC007278.2, AC007278.3, CYP2C19, IL18R1, IL18RAP, IL1RL1, RP11-400G3.5, SLC9A4, ZNF32
GERP++	μ = -1.007 [-9.62 – 4.57]	potentially regulated gene(s)	AC007278.2, AC007278.3, IL18R1, IL18RAP, IL1R2, IL1RL1, SLC9A2, SLC9A4
CADD score	μ = 2.505 [0.044 – 14.43]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Serum protein levels (sST2)	$<1.00\times10^{-35}$	GWAS Catalog	23999434	1
Inflammatory bowel disease	$<3.00\times10^{-20}$	GWAS Catalog	23128233	1
Atopic dermatitis	$<8.00\times10^{-18}$	GWAS Catalog	23042114	1
Crohn's disease (time to surgery)	$<2.00\times10^{-12}$	GWAS Catalog	21102463	1
CELIAC DISEASE	$<1.00\times10^{-15}$	GWAS Catalog	20190752	1
CELIAC DISEASE	$<4.00\times10^{-9}$	GWAS Catalog	18311140	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL18R1	?	ENSG00000115604	caudate basal ganglia	4.85×10^{-7} (p-value)	GTEx Portal V6	6
IL1RL1	?	ENSG00000115602	caudate basal ganglia	3.05×10^{-6} (p-value)	GTEx Portal V6	1
IL18R1	?	ENSG00000115604	lung	1.25×10^{-7} (p-value)	GTEx Portal V6	93
IL1RL1	?	ENSG00000115602	lung	1.32×10^{-8} (p-value)	GTEx Portal V6	92
AC007278.3	?	ENSG00000234389	lung	1.51×10^{-7} (p-value)	GTEx Portal V6	81
IL18RAP	?	ENSG00000115607	lung	9.04×10^{-6} (p-value)	GTEx Portal V6	1
AC007248.7	?	ENSG00000236785	lung	1.19×10^{-5} (p-value)	GTEx Portal V6	2
AC007278.2	?	ENSG00000236525	blood	1.05×10^{-13} (p-value)	GTEx Portal V6	93
IL18RAP	?	ENSG00000115607	blood	4.09×10^{-31} (p-value)	GTEx Portal V6	93
AC007278.3	?	ENSG00000234389	blood	9.36×10^{-17} (p-value)	GTEx Portal V6	93
IL18R1	?	ENSG00000115604	blood	4.67×10^{-6} (p-value)	GTEx Portal V6	38

IL18R1	?	ENSG00000115604	skeletal muscle	7.19×10-7 (p-value)	GTEx Portal V6	92
SLC9A4	?	ENSG00000180251	testis	2.01×10-7 (p-value)	GTEx Portal V6	92
IL18R1	?	ENSG00000115604	sun exposed skin	9.57×10-10 (p-value)	GTEx Portal V6	92
IL18R1	?	ENSG00000115604	left ventricle	2.14×10-6 (p-value)	GTEx Portal V6	55
IL18R1	?	ENSG00000115604	hippocampus	4.34×10-7 (p-value)	GTEx Portal V6	88
IL18R1	?	ENSG00000115604	cerebellum	1.83×10-6 (p-value)	GTEx Portal V6	30
IL18R1	ENST00000409599	ILMN_1781700	blood	6.65×10-5 (p-value)	MuTHER consortium	1
IL18R1	ENST00000233957					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ZNF32	ENST00000395797	ILMN_1680072	chr10	blood	1.31×10-8 (p-value)	Westra et al.	3
ZNF32	ENST00000374433						
CYP2C19	ENST00000371321	ILMN_1729191	chr10	blood	1.18×10-13 (p-value)	Westra et al.	3
RP11-400G3.5	ENST00000464755						

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000493988	1	ENST00000410040	IL18R1
		ENST00000404917, ENST00000473175, ENST00000447231, ENST00000233954	IL1RL1
FFCP00000494102	1	ENST00000409369, ENST00000450855	IL18RAP

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000027320		
ENCP00000027322		
ENCP00000027323		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000257210	2	ENCP00000027298	IL1R2
		ENCP00000027315	IL18R1 IL1RL1
		ENCP00000027324	AC007278.3
		ENCP00000027320	IL18RAP
		ENCP00000027322	IL18RAP
		ENCP00000027326	SLC9A4
		ENCP00000027328	SLC9A2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001655316 (CTCF binding site)	2	monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
ENSR00001045581 (CTCF binding site)	1	HSMMtube	H3K27me3
		blood (K562)	H3K27me3
ENSR00000303075 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1
		HSMMtube	H3K27me3
		A549	H3K27me3

		blood (K562)	Ini1, DNase1, H2AZ, H3K4me3
ENSR00001655328 (enhancer)	2	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K4me1
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001655330 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27me3
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	DNase1
		blood (GM12878)	BATF, NFKB, H3K4me2
		A549	H3K27me3
ENSR00000595668 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	DNase1
		blood (GM12878)	BATF, NFKB, H3K4me2
		A549	H3K27me3
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
ENSR00000677145 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF, H3K27me3
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF, Rad21
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	CTCF
		A549	CTCF
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

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC007248.7	upstream gene variant, downstream gene variant	600	ENST00000430344	?	?	6
AC007278.2	upstream gene variant, downstream gene variant	406	ENST00000436582	?	?	10
AC007278.3	upstream gene variant, downstream gene variant	152	ENST00000450893	?	?	10
IL18R1	upstream gene variant, downstream gene variant	524	ENST00000409599	?	ENSP00000387211	2
IL18R1	downstream gene variant	740	ENST00000334376	?	ENSP00000334030	5
IL18R1	upstream gene variant, downstream gene variant	263	ENST00000410040	?	ENSP00000386663	4
IL18R1	upstream gene variant, downstream gene variant	640	ENST00000466357	?	?	5
IL18R1	upstream gene variant, downstream gene variant	878	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	2
IL18RAP	upstream gene variant, downstream gene variant	32	ENST00000497795	?	?	5
IL18RAP	upstream gene variant, downstream gene variant	105	ENST00000264260	NM_003853.2	ENSP00000264260	5
IL18RAP	upstream gene variant, downstream gene variant	861	ENST00000450855	?	ENSP00000389815	3
IL18RAP	upstream gene variant, downstream gene variant	1102	ENST00000409369	?	ENSP00000387201	5
IL1RL1	upstream gene variant	1867	ENST00000409584	?	ENSP00000386618	1
IL1RL1	upstream gene variant	1859	ENST00000463990	?	?	1
IL1RL1	upstream gene variant	302	ENST00000447231	?	ENSP00000409437	3
IL1RL1	upstream gene variant	1890	ENST00000482701	?	?	1
IL1RL1	upstream gene variant	1897	ENST00000427077	?	ENSP00000391120	1
IL1RL1	upstream gene variant, downstream gene variant	1757	ENST00000311734	NM_003856.3	ENSP00000310371	3
IL1RL1	upstream gene variant, downstream gene variant	236	ENST00000233954	NM_016232.4	ENSP00000233954	4
IL1RL1	upstream gene variant	263	ENST00000473175	?	?	3
IL1RL1	upstream gene variant, downstream gene variant	236	ENST00000404917	NM_001282408.1	ENSP00000384822	4
MIR4772	upstream gene variant, downstream gene variant	282	ENST00000581495	?	?	10
SLC9A4	upstream gene variant	2993	ENST00000295269	NM_001011552.3	ENSP00000295269	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC007278.2	ENST00000436582	?	?	2
IL18R1	ENST00000233957	NM_003855.3, NM_001282399.1	ENSP00000233957	22
IL18R1	ENST00000334376	?	ENSP00000334030	11
IL18R1	ENST00000410040	?	ENSP00000386663	27
IL18R1	ENST00000409599	?	ENSP00000387211	22
IL18RAP	ENST00000409369	?	ENSP00000387201	26
IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	26

IL18RAP	ENST00000264260	NM_003853.2	ENSP00000264260	26
IL18RAP	ENST00000450855	?	ENSP00000389815	5
IL18RAP	ENST00000497795	?	?	2
IL1RL1	ENST00000404917	NM_001282408.1	ENSP00000384822	3
IL1RL1	ENST00000447231	?	ENSP00000409437	3
IL1RL1	ENST00000233954	NM_016232.4	ENSP00000233954	4
IL1RL1	ENST00000473175	?	?	3
SLC9A4	ENST00000295269	NM_001011552.3	ENSP00000295269	5

3'-UTR variant

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

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC007278.2	ENST00000436582	?	1
AC007278.3	ENST00000450893	?	1