Locus 1 (rs5498)

SNiPAcard

Block annotations

Block info

genomic range	chr19:10,391,770-10,409,323
block size	17,554 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.064 [-2.744 – 0.21]	gene(s) hit or close-by	CTD-2369P2.5, CTD-2369P2.8, ICAM1, ICAM4, ICAM5
phastCons	μ = 0.040 [0 – 0.256]	eQTL gene(s)	ICAM4, ICAM5
GERP++	μ = -0.089 [-2.57 – 2.66]	potentially regulated gene(s)	ICAM4, KRI1, P2RY11, PPAN, TYK2, TYK2, ZGLP1
CADD score	μ = 4.307 [0.068 – 10.49]	disease gene(s)	ICAM1, ICAM4, TYK2, P2RY11

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Soluble ICAM-1	$<6.00\times10^{-89}$	GWAS Catalog	21533024	1
Soluble levels of adhesion molecules	$<4.00\times10^{-23}$	GWAS Catalog	20167578	1
Soluble ICAM-1	$<5.00\times10^{-25}$	GWAS Catalog	18604267	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM003891	1

Disease gene annotation

gene	trait	source DB	source entry/link
ICAM1	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
ICAM4	BLOOD GROUP SYSTEM, LANDSTEINER-WIENER	OMIM	MIM:111250
TYK2	IMMUNODEFICIENCY 35	OMIM	MIM:611521
ICAM1	Chronic mucocutaneous candidosis	OrphaNet	OrphaNet:1334
TYK2	Oligoarticular juvenile arthritis	OrphaNet	OrphaNet:85410
TYK2	Juvenile rheumatoid factor-negative polyarthritis	OrphaNet	OrphaNet:85408
TYK2	Autosomal recessive hyper-IgE syndrome due to TYK2 deficiency	OrphaNet	OrphaNet:331226
P2RY11	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
ICAM1	missense variant	ENST00000423829	?	ENSP00000413124	E/K	Gag/Aag	?	?	1
ICAM1	missense variant	ENST00000264832	NM_000201.2	ENSP00000264832	E/K	Gag/Aag	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ICAM5	?	ENSG00000105376	transformed fibroblasts	3.11×10^{-10} (p-value)	GTEx Portal V6	11
ICAM5	?	ENSG00000105376	thyroid	1.35×10^{-7} (p-value)	GTEx Portal V6	11
ICAM4	?	ENSG00000105371	blood	1.55×10^{-8} (p-value)	GTEx Portal V6	10
ICAM4	ENST00000380770	ILMN_1681296	blood	1.21×10^{-26} (p-value)	MuTHER consortium	4
ICAM4	ENST00000393717					
ICAM4	ENST00000340992					
ICAM4	ENST00000380770	ILMN_1681296	monocyte	5.86×10^{-18} (p-value)	Fairfax et al.	3
ICAM4	ENST00000393717					
ICAM4	ENST00000340992					
?	?	ILMN_2393067	monocyte	2.44×10^{-14} (p-value)	Fairfax et al.	3

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000309935	1	ENST00000380770, ENST00000393717, ENST00000340992	ICAM4

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000223642	1	ENCP00000024048	TYK2 TYK2
		ENCP00000024047	TYK2 TYK2
		ENCP00000024036	PPAN
		ENCP00000024049	TYK2 TYK2
		ENCP00000024035	PPAN
		ENCP00000024037	P2RY11
		ENCP00000024051	KRI1
		ENCP00000024034	PPAN
		ENCP00000024038	ZGLP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001417842 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me2, SP1, USF1, H3K4me3, H3K27me3, H3K36me3
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	H3K4me3, DNase1, H3K36me3, Egr1, H3K27ac, Max, PU1, E2F6, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, CTCF
		skin (NHDF-AD)	H3K4me2, H3K27me3
		muscle (HSMM)	H2AZ, H3K4me2, H3K27me3
		liver (HepG2)	H3K4me1, H3K4me2, H3K4me3, H3K36me3, H3K27me3, DNase1
		blood (GM12878)	DNase1, PU1, PolII, H2AZ, CTCF, Egr1, BCL3, H3K4me3, H3K4me2, H3K36me3
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3
		nervous (NH-A)	DNase1, H3K4me2, H3K27me3
		skin (NHEK)	H3K27me3, H3K4me3, H3K4me2
		NHLF	H3K27me3
		Osteobl	H3K4me2, H3K27me3
		blood (DND-41)	H3K36me3, H3K4me1
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, PolII, H3K4me3, Max, H3K27ac, H3K4me2, H3K9ac, Cmyc, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H4K20me1, H3K36me3, H3K27me3, H3K4me3

		endothelium (HUVEC) A549	H3K36me3, DNase1, H3K27me3, Max DNase1, H3K4me2, H3K4me3, H3K27me3
ENSR00000343265 e! (promoter flanking region)	2	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	CTCF, Rad21, H3K4me2, H3K27me3, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	PolII, H3K36me3
		skin (NHDF-AD)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3, H3K4me1, DNase1
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	H3K4me2, H3K4me3
		blood (GM12878)	PolII, H2AZ, ELF1, H3K4me3, DNase1
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K27me3
		A549	H3K4me2, H3K4me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTD-2369P2.5 e!	upstream gene variant	1439	ENST00000592893 e!	?	?	2
CTD-2369P2.8 e!	downstream gene variant, upstream gene variant	794	ENST00000589379 e!	?	?	5
ICAM1 e!	upstream gene variant, downstream gene variant	724	ENST00000592686 e!	?	?	6
ICAM1 e!	downstream gene variant	1254	ENST00000588645 e!	?	ENSP00000465680 e!	3
ICAM1 e!	upstream gene variant, downstream gene variant	296	ENST00000585443 e!	?	?	5
ICAM1 e!	downstream gene variant	112	ENST00000264832 e!	NM_000201.2	ENSP00000264832 e!	2
ICAM1 e!	downstream gene variant	964	ENST00000423829 e!	?	ENSP00000413124 e!	3
ICAM4 e!	upstream gene variant, downstream gene variant	247	ENST00000340992 e!	NM_001039132.2	ENSP00000342114 e!	5
ICAM4 e!	upstream gene variant, downstream gene variant	277	ENST00000393717 e!	NM_022377.3	ENSP00000377320 e!	5
ICAM4 e!	upstream gene variant, downstream gene variant	240	ENST00000380770 e!	NM_001544.4	ENSP00000370147 e!	5
ICAM5 e!	upstream gene variant, downstream gene variant	866	ENST00000586480 e!	?	?	5
ICAM5 e!	upstream gene variant, downstream gene variant	555	ENST00000587398 e!	?	ENSP00000464796 e!	5
ICAM5 e!	upstream gene variant, downstream gene variant	3266	ENST00000588912 e!	?	?	4
ICAM5 e!	upstream gene variant, downstream gene variant	3297	ENST00000586004 e!	?	?	4
ICAM5 e!	upstream gene variant, downstream gene variant	864	ENST00000221980 e!	NM_003259.3	ENSP00000221980 e!	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2369P2.5 e!	ENST00000592893 e!	?	?	3
ICAM1 e!	ENST00000423829 e!	?	ENSP00000413124 e!	3
ICAM1 e!	ENST00000264832 e!	NM_000201.2	ENSP00000264832 e!	3
ICAM1 e!	ENST00000588645 e!	?	ENSP00000465680 e!	3
ICAM1 e!	ENST00000585443 e!	?	?	1
ICAM5 e!	ENST00000221980 e!	NM_003259.3	ENSP00000221980 e!	3
ICAM5 e!	ENST00000586004 e!	?	?	1
ICAM5 e!	ENST00000586480 e!	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ICAM1	ENST00000264832	NM_000201.2	ENSP00000264832	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTD-2369P2.5	ENST00000592893	?	1
CTD-2369P2.8	ENST00000589379	?	2
ICAM5	ENST00000588912	?	1