Locus 351 (rs1051738)

SNiPAcard

Block annotations

Block info

genomic range	chr19:10,547,226-10,591,142
block size	43,917 bp
variant count	22 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.580 [-2.858 – 0.464]	gene(s) hit or close-by	PDE4A
phastCons	μ = 0.023 [0 – 0.128]	eQTL gene(s)	ICAM4, KEAP1, S1PR5, SLC44A2, TYK2
GERP++	μ = -0.510 [-4.39 – 0.392]	potentially regulated gene(s)	KRI1, P2RY11, PPAN, PPAN-P2RY11, TYK2, TYK2, ZGLP1
CADD score	μ = 2.559 [0.035 – 10.39]	disease gene(s)	TYK2, ICAM4, P2RY11

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TYK2	IMMUNODEFICIENCY 35	OMIM	MIM:611521
ICAM4	BLOOD GROUP SYSTEM, LANDSTEINER-WIENER	OMIM	MIM:111250
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)
PDE4A	missense variant	ENST00000380702	NM_001111307.1	ENSP00000370078	E/A	gAg/gCg	?	?	1
PDE4A	missense variant	ENST00000344979	NM_006202.2	ENSP00000341007	E/A	gAg/gCg	?	?	1
PDE4A	missense variant	ENST00000440014	NM_001111309.1	ENSP00000394754	E/A	gAg/gCg	?	?	1
PDE4A	missense variant	ENST00000293683	NM_001111308.1	ENSP00000293683	E/A	gAg/gCg	?	?	1
PDE4A	missense variant	ENST00000592685	NM_001243121.1	ENSP00000468507	E/A	gAg/gCg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TYK2	?	ENSG00000105397	breast	4.56×10^{-6} (p-value)	GTEx Portal V6	1
KEAP1	ENST00000590237	ILMN_2410771	blood	7.39×10^{-6} (p-value)	Westra et al.	1
KEAP1	ENST00000592478					
KEAP1	ENST00000590593					
KEAP1	ENST00000393623					
KEAP1	ENST00000171111					

ICAM4	ENST00000380770	ILMN_1681296	blood	7.31×10-6 (p-value)	Westra et al.	1
ICAM4	ENST00000393717					
ICAM4	ENST00000340992					
KEAP1	ENST00000592478	ILMN_2410772	blood	1.09×10-5 (p-value)	Westra et al.	1
KEAP1	ENST00000590237					
KEAP1	ENST00000590593					
KEAP1	ENST00000171111					
KEAP1	ENST00000393623					
SLC44A2	ENST00000589561	ILMN_1771987	blood	5.04×10-5 (p-value)	Westra et al.	1
SLC44A2	ENST00000586078					
SLC44A2	ENST00000586549					
SLC44A2	ENST00000407327					
SLC44A2	ENST00000335757					
S1PR5	ENST00000333430	ILMN_2073184	blood	1.07×10-4 (p-value)	Westra et al.	1
S1PR5	ENST00000617721					
S1PR5	ENST00000439028					
ICAM4	ENST00000380770	ILMN_1681296	monocyte	5.54×10-4 (p-value)	Fairfax et al.	1
ICAM4	ENST00000393717					
ICAM4	ENST00000340992					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000223450	1	ENCP00000024048	TYK2 TYK2
		ENCP00000024033	PPAN
		ENCP00000024032	PPAN
		ENCP00000024047	TYK2 TYK2
		ENCP00000024049	TYK2 TYK2
		ENCP00000024036	PPAN
		ENCP00000024035	PPAN
		ENCP00000024037	P2RY11
		ENCP00000024031	PPAN-P2RY11 PPAN
		ENCP00000024051	KRI1
		ENCP00000024034	PPAN
		ENCP00000024038	ZGLP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000343297 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	H3K36me3
		Osteobl	CTCF
		blood (DND-41)	CTCF

breast (HMEC)	CTCF
cervix (HeLa-S3)	CTCF
monocytes (Monocytes-CD14+)	DNase1, CTCF
endothelium (HUVEC)	H3K36me3, CTCF
A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDE4A	downstream gene variant	327	ENST00000380702	NM_001111307.1	ENSP00000370078	5
PDE4A	downstream gene variant	174	ENST00000586275	?	?	5
PDE4A	downstream gene variant	2337	ENST00000293683	NM_001111308.1	ENSP00000293683	1
PDE4A	downstream gene variant	1211	ENST00000591971	?	ENSP00000467137	5
PDE4A	upstream gene variant, downstream gene variant	1783	ENST00000590407	?	?	3
PDE4A	downstream gene variant	2337	ENST00000440014	NM_001111309.1	ENSP00000394754	1
PDE4A	downstream gene variant	2593	ENST00000589073	?	ENSP00000465792	1
PDE4A	downstream gene variant	2027	ENST00000344979	NM_006202.2	ENSP00000341007	1
PDE4A	downstream gene variant	2309	ENST00000592685	NM_001243121.1	ENSP00000468507	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDE4A	ENST00000591971	?	ENSP00000467137	4
PDE4A	ENST00000293683	NM_001111308.1	ENSP00000293683	11
PDE4A	ENST00000344979	NM_006202.2	ENSP00000341007	9
PDE4A	ENST00000380702	NM_001111307.1	ENSP00000370078	11
PDE4A	ENST00000440014	NM_001111309.1	ENSP00000394754	11
PDE4A	ENST00000592685	NM_001243121.1	ENSP00000468507	11
PDE4A	ENST00000589073	?	ENSP00000465792	9
PDE4A	ENST00000586275	?	?	2
PDE4A	ENST00000590407	?	?	7

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDE4A	ENST00000589073	?	ENSP00000465792	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PDE4A	ENST00000590407	?	1