Locus 372 (rs2239805)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,380,208-32,428,862 e!
block size	48,655 bp
variant count	71 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.425 [-2.801 – 2.075]	**gene(s) hit or close-by**	HLA-DRA e!, HLA-DRB9 e!
phastCons	μ = 0.034 [0 – 0.454]	**eQTL gene(s)**	APOM e!, C4A e!, C4B e!, C6orf25 e!, CYP21A1P e!, CYP21A2 e!, HCP5 e!, HLA-DMA e!, HLA-DMB e!, HLA-DOB e!, HLA-DPB1 e!, HLA-DQA2 e!, HLA-DRA e!, HLA-DRB1 e!, HLA-DRB5 e!, HLA-DRB6 e!, HLA-DRB9 e!, LIMS1 e!, LST1 e!, LY6G6C e!, MICB e!, NOTCH4 e!, PSMB9 e!, RNF5 e!, TNXA e!, XXbac-BPG246D15.9 e!, XXbac-BPG300A18.13 e!
GERP++	μ = -0.137 [-3.41 – 3.16]	**potentially regulated gene(s)**	BTNL2 e!, BTNL2 e!, BTNL2 e!, BTNL2 e!, BTNL2 e!, BTNL2 e!, BTNL2 e!, BTNL2 e!, C4B e!, C4B e!, C4B e!, C4B e!, C4B e!, C4B e!, CYP21A2 e!, CYP21A2 e!, CYP21A2 e!, CYP21A2 e!, CYP21A2 e!, CYP21A2 e!, FKBPL e!, FKBPL e!, FKBPL e!, FKBPL e!, HLA-DOB e!, HLA-DOB e!, HLA-DOB e!, HLA-DOB e!, HLA-DOB e!, HLA-DOB e!, HLA-DQA1 e!, HLA-DQA1 e!, HLA-DQA1 e!, HLA-DQA1 e!, HLA-DQA1 e!, HLA-DQA1 e!, HLA-DQB1 e!, HLA-DQB1 e!, HLA-DQB1 e!, HLA-DQB1 e!, HLA-DQB1 e!, HLA-DQB1 e!, HLA-DQB1 e!, HLA-DQB2 e!, HLA-DQB2 e!, HLA-DQB2 e!, HLA-DQB2 e!, HLA-DQB2 e!, HLA-DQB2 e!, HLA-DQB2 e!, HLA-DQB2 e!, HLA-DRA e!, HLA-DRB5 e!, PBX2 e!, PBX2 e!, PBX2 e!, PBX2 e!, PBX2 e!, PBX2 e!, PBX2 e!, PRRT1 e!, PRRT1 e!, PRRT1 e!, PRRT1 e!, PRRT1 e!, PRRT1 e!, PRRT1 e!, PRRT1 e!, PSMB9 e!, PSMB9 e!, PSMB9 e!, PSMB9 e!, PSMB9 e!, PSMB9 e!, PSMB9 e!, PSMB9 e!, TAP1 e!, TAP1 e!, TAP1 e!, TAP1 e!, TAP1 e!, TAP1 e!, TAP1 e!, TAP1 e!
CADD score	μ = 3.146 [0.008 – 11.41]	**disease gene(s)**	C4B e!, C4A e!, CYP21A2 e!, HLA-DRA e!, e!, e!, e!, e!, e!, e!, e!, BTNL2 e!, e!, e!, e!, e!, e!, e!, e!, e!, e!, e!, e!, e!, TAP1 e!, e!, e!, HLA-DPB1 e!, HLA-DRB1 e!, HLA-DQB1 e!

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Lupus erythematosus, systemic	6.53×10^{-6}	dbGaP	pha002867	1
Lupus erythematosus, systemic	3.21×10^{-14}	dbGaP	pha002848	3

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CS024164	1

Disease gene annotation

gene	trait	source DB	source entry/link
C4B e!	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379
C4B e!	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4B e!	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A e!	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A e!	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A e!	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A e!	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
CYP21A2 e!	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910
HLA-DRA e!	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
e!	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
e!	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387

Locus 372 (sentinel SNP rs2239805 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
BTNL2	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
TAP1	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	OrphaNet	OrphaNet:315306	orphanet
CYP21A2	Nonclassic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	OrphaNet	OrphaNet:95698	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	OrphaNet	OrphaNet:315311	orphanet
HLA-DPB1	Chronic berylliosis	OrphaNet	OrphaNet:133	orphanet
HLA-DPB1	Granulomatosis with polyangiitis	OrphaNet	OrphaNet:900	orphanet
HLA-DRB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DRB1	Limited systemic sclerosis	OrphaNet	OrphaNet:220407	orphanet
HLA-DRB1	Limited cutaneous systemic sclerosis	OrphaNet	OrphaNet:220402	orphanet
HLA-DRB1	Follicular lymphoma	OrphaNet	OrphaNet:545	orphanet
HLA-DRB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DRB1	Diffuse cutaneous systemic sclerosis	OrphaNet	OrphaNet:220393	orphanet
HLA-DRB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
HLA-DRB1	Sarcoidosis	OrphaNet	OrphaNet:797	orphanet
HLA-DRA	Graham Little-Piccardi-Lassueur syndrome	OrphaNet	OrphaNet:505	orphanet
BTNL2	Sarcoidosis	OrphaNet	OrphaNet:797	orphanet
HLA-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
TAP1	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NOTCH4	ENST00000474612	ILMN_1711157	adipocyte	1.12×10-10 (p-value)	MuTHER consortium	36
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
HLA-DPB1	ENST00000418931	ILMN_1749070	skin	4.45×10-9 (p-value)	MuTHER consortium	36
HLA-DPB1	ENST00000416804		blood	1.14×10-7 (p-value)	MuTHER consortium	36
			adipocyte	2.96×10-7 (p-value)	MuTHER consortium	36
MICB	ENST00000538442	ILMN_1708006	skin	1.36×10-8 (p-value)	MuTHER consortium	36
MICB	ENST00000399150					
MICB	ENST00000252229					
PSMB9	ENST00000467593	ILMN_1798233	blood	2.24×10-8 (p-value)	MuTHER consortium	36
PSMB9	ENST00000464863					
HLA-DMA	ENST00000395305	ILMN_1695311	blood	5.67×10-6 (p-value)	MuTHER consortium	32
HLA-DMA	ENST00000480785					
HLA-DMA	ENST00000464392					
HLA-DMA	ENST00000456800					
HLA-DMA	ENST00000395303					
HLA-DMA	ENST00000374843					
HLA-DMA	ENST00000477541					
HLA-DMB	ENST00000487996	ILMN_1761733	blood	3.21×10-5 (p-value)	MuTHER consortium	1
HLA-DMB	ENST00000498020					
HLA-DMB	ENST00000477537					
HLA-DMB	ENST00000418107					
HLA-DMB	ENST00000414017					
HLA-DMB	ENST00000438510					
HCP5	ENST00000414046	ILMN_1803945	adipocyte	3.50×10-5 (p-value)	MuTHER consortium	1
HCP5	ENST00000541196					
C4A	?	ENSG00000244731	muscularis mucosae	1.81×10-12 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	muscularis mucosae	2.03×10-6 (p-value)	GTEx Portal V6	10
C4A	?	ENSG00000244731	lung	1.13×10-9 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	lung	1.08×10-7 (p-value)	GTEx Portal V6	64
HLA-DRB6	?	ENSG00000229391	lung	4.18×10-6 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	atrial appendage	5.98×10-11 (p-value)	GTEx Portal V6	69
C4B	?	ENSG00000224389	transformed fibroblasts	4.39×10-8 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	transformed fibroblasts	9.62×10-14 (p-value)	GTEx Portal V6	69
C4B	?	ENSG00000224389	blood	4.64×10-10 (p-value)	GTEx Portal V6	67

C4A	?	ENSG00000244731	blood	3.57×10-11 (p-value)	GTEx Portal V6	69
HLA-DRB6	?	ENSG00000229391	blood	1.25×10-9 (p-value)	GTEx Portal V6	69
HLA-DQA2	?	ENSG00000237541	blood	3.38×10-8 (p-value)	GTEx Portal V6	19
HLA-DRB5	?	ENSG00000198502	blood	5.32×10-6 (p-value)	GTEx Portal V6	4
PSMB9	ENST00000467593	ILMN_1798233	monocyte	2.30×10-5 (p-value)	Fairfax et al.	20
PSMB9	ENST00000464863		b-cell	1.37×10-6 (p-value)	Fairfax et al.	20
HLA-DPB1	ENST00000418931	ILMN_1749070	monocyte	2.88×10-5 (p-value)	Fairfax et al.	18
HLA-DPB1	ENST00000416804		b-cell	3.20×10-6 (p-value)	Fairfax et al.	18
HLA-DOB	ENST00000438763	ILMN_1700428	monocyte	9.14×10-5 (p-value)	Fairfax et al.	19
HLA-DOB	ENST00000488325					
HLA-DOB	ENST00000475235					
XXbac-BPG246D15.9	ENST00000452392					
C4A	?	ENSG00000244731	thyroid	5.52×10-12 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	thyroid	7.18×10-13 (p-value)	GTEx Portal V6	69
RNF5	?	ENSG00000204308	thyroid	4.39×10-7 (p-value)	GTEx Portal V6	58
HLA-DRB1	?	ENSG00000196126	thyroid	1.01×10-5 (p-value)	GTEx Portal V6	3
HLA-DMA	?	ENSG00000204257	thyroid	3.20×10-9 (p-value)	GTEx Portal V6	69
CYP21A1P	?	ENSG00000204338	thyroid	5.40×10-9 (p-value)	GTEx Portal V6	63
HLA-DRB6	?	ENSG00000229391	thyroid	3.59×10-6 (p-value)	GTEx Portal V6	10
C4A	?	ENSG00000244731	skeletal muscle	4.15×10-9 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	skeletal muscle	6.15×10-10 (p-value)	GTEx Portal V6	69
HLA-DRB6	?	ENSG00000229391	skeletal muscle	7.72×10-7 (p-value)	GTEx Portal V6	61
APOM	?	ENSG00000204444	skeletal muscle	4.71×10-6 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	transverse colon	7.43×10-10 (p-value)	GTEx Portal V6	63
HLA-DMA	?	ENSG00000204257	sun exposed skin	3.28×10-7 (p-value)	GTEx Portal V6	52
C4B	?	ENSG00000224389	sun exposed skin	4.54×10-10 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	sun exposed skin	1.31×10-8 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	sun exposed skin	2.17×10-6 (p-value)	GTEx Portal V6	12
CYP21A2	?	ENSG00000231852	sun exposed skin	2.20×10-9 (p-value)	GTEx Portal V6	62
HLA-DRB6	?	ENSG00000229391	sun exposed skin	1.86×10-6 (p-value)	GTEx Portal V6	25
HLA-DRB9	?	ENSG00000196301	sun exposed skin	3.11×10-6 (p-value)	GTEx Portal V6	7
C4A	?	ENSG00000244731	unexposed skin	1.89×10-11 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	left ventricle	2.32×10-14 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	left ventricle	2.45×10-7 (p-value)	GTEx Portal V6	61

CYP21A1P	?	ENSG00000204338	left ventricle	9.13×10-9 (p-value)	GTEx Portal V6	69
NOTCH4	?	ENSG00000204301	left ventricle	7.47×10-8 (p-value)	GTEx Portal V6	61
C4A	?	ENSG00000244731	aorta	2.96×10-8 (p-value)	GTEx Portal V6	67
HLA-DRB5	?	ENSG00000198502	aorta	7.96×10-7 (p-value)	GTEx Portal V6	15
CYP21A2	?	ENSG00000231852	aorta	8.94×10-7 (p-value)	GTEx Portal V6	48
MICB	?	ENSG00000204516	aorta	3.81×10-7 (p-value)	GTEx Portal V6	7
C4A	?	ENSG00000244731	subcutaneous adipocytes	1.06×10-10 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	subcutaneous adipocytes	2.11×10-11 (p-value)	GTEx Portal V6	69
CYP21A2	?	ENSG00000231852	subcutaneous adipocytes	1.64×10-6 (p-value)	GTEx Portal V6	17
NOTCH4	?	ENSG00000204301	subcutaneous adipocytes	3.67×10-6 (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	stomach	3.64×10-13 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	visceral adipocytes	5.84×10-9 (p-value)	GTEx Portal V6	67
HLA-DRB5	?	ENSG00000198502	visceral adipocytes	7.47×10-8 (p-value)	GTEx Portal V6	66
C4A	?	ENSG00000244731	tibial nerve	1.49×10-13 (p-value)	GTEx Portal V6	69
XXbac-BPG300A18.13	?	ENSG00000273333	tibial nerve	4.12×10-6 (p-value)	GTEx Portal V6	38
C4A	?	ENSG00000244731	esophagus mucosa	2.22×10-12 (p-value)	GTEx Portal V6	69
HLA-DRB5	?	ENSG00000198502	esophagus mucosa	7.42×10-9 (p-value)	GTEx Portal V6	66
LY6G6C	?	ENSG00000204421	esophagus mucosa	1.01×10-6 (p-value)	GTEx Portal V6	58
C6orf25	?	ENSG00000204420	esophagus mucosa	7.14×10-8 (p-value)	GTEx Portal V6	57
MICB	?	ENSG00000204516	esophagus mucosa	6.07×10-6 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	gastroesophageal junction	1.45×10-7 (p-value)	GTEx Portal V6	41
?	?	221491_x_at	blood	1.80×10-21 (p-value)	Dixon et al.	3
?	?	215666_at	blood	4.10×10-9 (p-value)	Dixon et al.	2
C4A	?	ENSG00000244731	testis	1.97×10-11 (p-value)	GTEx Portal V6	69
C4A	?	ENSG00000244731	pancreas	6.53×10-7 (p-value)	GTEx Portal V6	45
C4A	?	ENSG00000244731	breast	1.35×10-8 (p-value)	GTEx Portal V6	62
C4A	?	ENSG00000244731	tibial artery	5.99×10-7 (p-value)	GTEx Portal V6	42
CYP21A1P	?	ENSG00000204338	tibial artery	5.54×10-7 (p-value)	GTEx Portal V6	46
HLA-DRB5	?	ENSG00000198502	tibial artery	4.29×10-7 (p-value)	GTEx Portal V6	6
HLA-DRB6	?	ENSG00000229391	tibial artery	7.53×10-6 (p-value)	GTEx Portal V6	4
C4A	?	ENSG00000244731	EBV lymphocytes	1.74×10-9 (p-value)	GTEx Portal V6	55
HLA-DRB5	?	ENSG00000198502	EBV lymphocytes	3.58×10-7 (p-value)	GTEx Portal V6	15

C4A	?	ENSG00000244731	liver	2.18×10-9 (p-value)	GTEx Portal V6	66
TNXA	?	ENSG00000248290	adrenal gland	1.46×10-8 (p-value)	GTEx Portal V6	67
HLA-DMA	?	ENSG00000204257	cerebellum	3.89×10-7 (p-value)	GTEx Portal V6	67
C4A	?	ENSG00000244731	cerebellum	1.80×10-7 (p-value)	GTEx Portal V6	50
?	?	ILMN_2159694	monocyte	3.60×10-21 (p-value)	Zeller et al.	9
?	?	ILMN_1893395	monocyte	2.44×10-20 (p-value)	Zeller et al.	9
?	?	ILMN_1752592	monocyte	8.41×10-62 (p-value)	Zeller et al.	9
HLA-DRA	ENST00000374982	ILMN_2157441	monocyte	2.49×10-20 (p-value)	Zeller et al.	9
HLA-DRA	ENST00000395388					
HLA-DRB6	ENST00000437183	ILMN_2066060	monocyte	1.15×10-33 (p-value)	Zeller et al.	9
HLA-DRB6	ENST00000411500					
HLA-DRB6	ENST00000437650					
PSMB9	ENST00000467593	ILMN_1798233	monocyte	1.82×10-12 (p-value)	Zeller et al.	1
PSMB9	ENST00000464863					
HLA-DOB	ENST00000438763	ILMN_1700428	monocyte	9.48×10-15 (p-value)	Zeller et al.	7
HLA-DOB	ENST00000475235					
HLA-DOB	ENST00000488325					
XXbac-BPG246D15.9	ENST00000452392					
?	?	ILMN_1715169	monocyte	4.42×10-44 (p-value)	Zeller et al.	9
HLA-DPB1	ENST00000418931	ILMN_1749070	monocyte	9.64×10-17 (p-value)	Zeller et al.	9
HLA-DPB1	ENST00000416804					
HLA-DRB5	ENST00000374975	ILMN_1697499	monocyte	3.78×10-17 (p-value)	Zeller et al.	9
?	?	ILMN_1883997	monocyte	2.12×10-26 (p-value)	Zeller et al.	9
LST1	ENST00000419073	ILMN_1717127	monocyte	1.49×10-13 (p-value)	Zeller et al.	2
LST1	ENST00000433492					
LST1	ENST00000376089					
LST1	ENST00000376090					
LST1	ENST00000211921					
HLA-DRB5	?	ENSG00000198502	brain	9.16×10-4 (q-value)	SeeQTL DB (Myers et al.)	7
C4A	?	ENSG00000244731	spleen	9.55×10-7 (p-value)	GTEx Portal V6	1
HLA-DRB5	?	ENSG00000198502	coronary artery	4.40×10-7 (p-value)	GTEx Portal V6	5
HLA-DRB5	?	ENSG00000198502	sigmoid colon	1.20×10-6 (p-value)	GTEx Portal V6	3

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
LIMS1	ENST00000410093	ILMN_1675387	chr2	monocyte	4.06×10-12 (p-value)	Zeller et al.	1
LIMS1	ENST00000542845						

LIMS1	ENST00000338045
LIMS1	ENST00000480744
LIMS1	ENST00000393310
LIMS1	ENST00000332345
LIMS1	ENST00000409441
LIMS1	ENST00000544547

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000739297	1	ENST00000374982, ENST00000395388	HLA-DRA

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445325	3	ENCP00000048170	FKBPL FKBPL FKBPL FKBPL
		ENCP00000048221	HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2 HLA-DQB2
ENCE00000445329	1	ENCP00000048200	BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2
		ENCP00000048171	PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1
ENCE00000445291	1	ENCP00000048233	PSMB9 PSMB9 PSMB9 PSMB9 PSMB9 PSMB9 PSMB9 PSMB9
		ENCP00000048222	HLA-DOB HLA-DOB HLA-DOB HLA-DOB HLA-DOB HLA-DOB
		ENCP00000048159	CYP21A2 CYP21A2 C4B CYP21A2 C4B C4B

			CYP21A2 C4B C4B CYP21A2 C4B CYP21A2
ENCE00000445363	2	ENCP00000048231	TAP1 TAP1 TAP1 TAP1 TAP1 TAP1 TAP1 TAP1
		ENCP00000048185	PBX2 PBX2 PBX2 PBX2 PBX2 PBX2 PBX2
		ENCP00000048216	HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1
		ENCP00000048209	HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1
		ENCP00000048215	HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1 HLA-DQB1
		ENCP00000048211	HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1 HLA-DQA1
		ENCP00000048204	HLA-DRB5
		ENCP00000048187	PBX2 PBX2 PBX2 PBX2 PBX2 PBX2 PBX2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000025344 (CTCF binding site)	9	embryonic stem cell (H1ESC)	H3K4me2, Rad21, CTCF, H3K4me3, DNase1
		HSMMtube	H3K27me3, CTCF
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF, H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		nervous (NH-A)	CTCF, H3K4me2
		skin (NHEK)	CTCF

		NHLF	CTCF
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K4me2, CTCF
		A549	H3K27me3, CTCF
ENSR00001703124 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K4me2, Rad21, CTCF, H3K4me3, DNase1
		HSMMtube	H3K27me3, CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF, H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF, H3K4me2
		skin (NHEK)	CTCF
		NHLF	CTCF, H3K27me3
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K27me3
		endothelium (HUVEC)	H3K4me2, CTCF, H3K27me3
		A549	H3K27me3, CTCF
ENSR00000488005 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		blood (K562)	H3K27me3
ENSR00000488009 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me2, SP1
		HSMMtube	H3K27me3
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	H3K4me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, Yy1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac, H3K36me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HLA-DRA	upstream gene variant, downstream gene variant	1044	ENST00000374982	?	ENSP00000364121	3
HLA-DRA	upstream gene variant, downstream gene variant	1008	ENST00000395388	NM_019111.4	ENSP00000378786	3
HLA-DRB9	downstream gene variant	1327	ENST00000449413	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HLA-DRA	ENST00000374982	?	ENSP00000364121	8
HLA-DRA	ENST00000395388	NM_019111.4	ENSP00000378786	8
HLA-DRB9	ENST00000449413	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
HLA-DRA	ENST00000374982	?	ENSP00000364121	1

HLA-DRA	ENST00000395388	NM_019111.4	ENSP00000378786	1