Locus 206 (rs1794282)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,218,120-32,776,921
block size	558,802 bp
variant count	30 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.450 [-3.618 – 0.444]	gene(s) hit or close-by	BTNL2, C6orf10, HLA-DOB, HLA-DRA, HLA-DRB9, HNRNPA1P2, RNU6-603P, XXbac-BPG154L12.4, XXbac-BPG154L12.5, XXbac-BPG246D15.9
phastCons	μ = 0.127 [0 – 0.923]	eQTL gene(s)	C4A, C4B, CYP21A1P, CYP21A2, HCP5, HLA-C, HLA-DMA, HLA-DMB, HLA-DPB1, HLA-DQA1, HLA-DQB1, HLA-DQB1-AS1, HLA-DQB2, HLA-DRB5, HLA-DRB6, LY6G6C, MICB, NOTCH4, PSMB9, RNF5, STK19, STK19B, TAP2, TNXA, TNXB, VARS2, WASF5P, XXbac-BPG246D15.9, XXbac-BPG248L24.13
GERP++	μ = -0.271 [-5.03 – 1.82]	potentially regulated gene(s)	BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, C2, C2, C2, C2, C2, C2, C2, C2, C2, C2, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, EGFL8, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HCG23, HLA-DRB6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6, XXbac-BPG254F23.6
CADD score	μ = 2.492 [0.026 – 15.24]	disease gene(s)	C4B, C4A, CYP21A2, VARS2, , , , , , , , , , C2, , , , , , , BTNL2, HLA-DRA, TAP2, TNXB, HLA-DPB1, HLA-DQB1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Lupus erythematosus, systemic	2.21×10^{-7}	dbGaP	pha002867	dbGaP	1
Diabetes mellitus, type 1	2.48×10^{-103}	dbGaP	pha002862	dbGaP	1
Lupus erythematosus, systemic	4.01×10^{-18}	dbGaP	pha002848	dbGaP	1

Disease gene annotation

gene	trait	source DB	source entry/link	
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379	OMIM
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380	OMIM
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790	OMIM
CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910	OMIM
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM

	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
BTNL2	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
HLA-DRA	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
TAP2	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571	OMIM
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839	orphanet
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285	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
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	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
HLA-DRA	Graham Little-Piccardi-Lassueur syndrome	OrphaNet	OrphaNet:505	orphanet
TAP2	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)
C4A	?	ENSG00000244731	pancreas	5.71×10^{-13} (p-value)	GTEx Portal V6	24
RNF5	?	ENSG00000204308	pancreas	1.41×10^{-6} (p-value)	GTEx Portal V6	22
XXbac-BPG248L24.13	?	ENSG00000256166	pancreas	3.28×10^{-7} (p-value)	GTEx Portal V6	5
HLA-DMA	?	ENSG00000204257	pancreas	2.02×10^{-11} (p-value)	GTEx Portal V6	24
WASF5P	?	ENSG00000231402	pancreas	4.68×10^{-7} (p-value)	GTEx Portal V6	5
CYP21A1P	?	ENSG00000204338	pancreas	2.84×10^{-7} (p-value)	GTEx Portal V6	14
HLA-DQB2	?	ENSG00000232629	muscularis mucosae	2.71×10^{-7} (p-value)	GTEx Portal V6	19
C4A	?	ENSG00000244731	muscularis mucosae	2.15×10^{-19} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	muscularis mucosae	3.72×10^{-7} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	muscularis mucosae	2.74×10^{-12} (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	lung	7.65×10^{-7} (p-value)	GTEx Portal V6	20

C4A	?	ENSG00000244731	lung	4.85×10-17 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	lung	1.72×10-8 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	lung	5.42×10-10 (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	atrial appendage	6.02×10-8 (p-value)	GTEx Portal V6	19
C4A	?	ENSG00000244731	atrial appendage	6.06×10-16 (p-value)	GTEx Portal V6	25
CYP21A1P	?	ENSG00000204338	atrial appendage	1.27×10-9 (p-value)	GTEx Portal V6	25
HLA-DMA	?	ENSG00000204257	transformed fibroblasts	1.88×10-20 (p-value)	GTEx Portal V6	24
HLA-DMB	?	ENSG00000242574	transformed fibroblasts	5.14×10-13 (p-value)	GTEx Portal V6	21
C4B	?	ENSG00000224389	transformed fibroblasts	1.54×10-15 (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	transformed fibroblasts	2.95×10-17 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	transformed fibroblasts	3.17×10-8 (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	transformed fibroblasts	9.47×10-6 (p-value)	GTEx Portal V6	1
HLA-DMA	?	ENSG00000204257	tibial artery	2.07×10-9 (p-value)	GTEx Portal V6	20
C4A	?	ENSG00000244731	tibial artery	2.30×10-13 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	tibial artery	7.01×10-10 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	tibial artery	1.84×10-12 (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	breast	1.15×10-14 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	breast	9.99×10-10 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	breast	6.62×10-13 (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	breast	1.26×10-7 (p-value)	GTEx Portal V6	10
STK19B	?	ENSG00000250535	breast	6.19×10-7 (p-value)	GTEx Portal V6	1
C4B	?	ENSG00000224389	blood	3.97×10-17 (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	blood	2.06×10-16 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	blood	3.69×10-8 (p-value)	GTEx Portal V6	4
CYP21A2	?	ENSG00000231852	blood	3.10×10-7 (p-value)	GTEx Portal V6	23
CYP21A1P	?	ENSG00000204338	blood	8.43×10-16 (p-value)	GTEx Portal V6	25
HLA-DMA	?	ENSG00000204257	thyroid	1.77×10-18 (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	thyroid	4.56×10-21 (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	thyroid	1.36×10-6 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	thyroid	2.70×10-18 (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	thyroid	9.43×10-7 (p-value)	GTEx Portal V6	12
HLA-DRB6	?	ENSG00000229391	thyroid	6.75×10-6 (p-value)	GTEx Portal V6	3
HLA-DQA1	?	ENSG00000196735	skeletal muscle	6.40×10-8 (p-value)	GTEx Portal V6	24
C4A	?	ENSG00000244731	skeletal muscle	5.08×10-20 (p-value)	GTEx Portal V6	25
HLA-DRB5	?	ENSG00000198502	skeletal muscle	6.10×10-7 (p-value)	GTEx Portal V6	20
HLA-C	?	ENSG00000204525	skeletal muscle	1.19×10-13 (p-value)	GTEx Portal V6	4
RNF5	?	ENSG00000204308	skeletal muscle	4.88×10-10 (p-value)	GTEx Portal V6	24

HLA-DRB6	?	ENSG00000229391	skeletal muscle	3.08×10^{-8} (p-value)	GTEx Portal V6	22
CYP21A1P	?	ENSG00000204338	skeletal muscle	6.94×10^{-7} (p-value)	GTEx Portal V6	20
HLA-DQB2	?	ENSG00000232629	skeletal muscle	1.40×10^{-6} (p-value)	GTEx Portal V6	9
HLA-DQB1-AS1	?	ENSG00000223534	skeletal muscle	2.02×10^{-6} (p-value)	GTEx Portal V6	7
HLA-DMA	?	ENSG00000204257	skeletal muscle	5.10×10^{-10} (p-value)	GTEx Portal V6	9
C4A	?	ENSG00000244731	transverse colon	1.71×10^{-12} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	transverse colon	6.93×10^{-7} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	transverse colon	1.98×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	EBV lymphocytes	9.01×10^{-10} (p-value)	GTEx Portal V6	21
HLA-C	?	ENSG00000204525	EBV lymphocytes	3.18×10^{-6} (p-value)	GTEx Portal V6	4
C4A	?	ENSG00000244731	unexposed skin	4.36×10^{-16} (p-value)	GTEx Portal V6	25
CYP21A2	?	ENSG00000231852	unexposed skin	9.86×10^{-7} (p-value)	GTEx Portal V6	18
CYP21A1P	?	ENSG00000204338	unexposed skin	5.73×10^{-10} (p-value)	GTEx Portal V6	24
MICB	?	ENSG00000204516	unexposed skin	2.73×10^{-6} (p-value)	GTEx Portal V6	16
HLA-DMA	?	ENSG00000204257	unexposed skin	1.94×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	sun exposed skin	4.91×10^{-17} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	sun exposed skin	3.99×10^{-8} (p-value)	GTEx Portal V6	4
NOTCH4	?	ENSG00000204301	sun exposed skin	2.52×10^{-9} (p-value)	GTEx Portal V6	21
HLA-DMA	?	ENSG00000204257	sun exposed skin	1.82×10^{-15} (p-value)	GTEx Portal V6	25
C4B	?	ENSG00000224389	sun exposed skin	8.56×10^{-14} (p-value)	GTEx Portal V6	24
CYP21A2	?	ENSG00000231852	sun exposed skin	1.11×10^{-10} (p-value)	GTEx Portal V6	25
MICB	?	ENSG00000204516	sun exposed skin	5.07×10^{-8} (p-value)	GTEx Portal V6	21
CYP21A1P	?	ENSG00000204338	sun exposed skin	2.27×10^{-13} (p-value)	GTEx Portal V6	25
HLA-DMA	?	ENSG00000204257	liver	4.11×10^{-13} (p-value)	GTEx Portal V6	25
STK19B	?	ENSG00000250535	liver	4.66×10^{-8} (p-value)	GTEx Portal V6	21
C4A	?	ENSG00000244731	liver	3.14×10^{-14} (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	left ventricle	7.06×10^{-8} (p-value)	GTEx Portal V6	21
C4A	?	ENSG00000244731	left ventricle	6.97×10^{-17} (p-value)	GTEx Portal V6	25
CYP21A1P	?	ENSG00000204338	left ventricle	1.10×10^{-9} (p-value)	GTEx Portal V6	25
NOTCH4	?	ENSG00000204301	left ventricle	7.25×10^{-8} (p-value)	GTEx Portal V6	24
C4A	?	ENSG00000244731	aorta	7.08×10^{-16} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	aorta	2.49×10^{-7} (p-value)	GTEx Portal V6	4
CYP21A2	?	ENSG00000231852	aorta	1.41×10^{-6} (p-value)	GTEx Portal V6	12
CYP21A1P	?	ENSG00000204338	aorta	1.34×10^{-8} (p-value)	GTEx Portal V6	24
MICB	?	ENSG00000204516	aorta	1.44×10^{-8} (p-value)	GTEx Portal V6	22
NOTCH4	?	ENSG00000204301	aorta	3.98×10^{-6} (p-value)	GTEx Portal V6	1
HLA-DQB2	?	ENSG00000232629	subcutaneous adipocytes	5.08×10^{-11} (p-value)	GTEx Portal V6	24

C4A	?	ENSG00000244731	subcutaneous adipocytes	1.31×10^{-17} (p-value)	GTEx Portal V6	25
HLA-DRB5	?	ENSG00000198502	subcutaneous adipocytes	3.29×10^{-7} (p-value)	GTEx Portal V6	23
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	1.50×10^{-8} (p-value)	GTEx Portal V6	4
NOTCH4	?	ENSG00000204301	subcutaneous adipocytes	3.45×10^{-7} (p-value)	GTEx Portal V6	21
C4B	?	ENSG00000224389	subcutaneous adipocytes	2.32×10^{-6} (p-value)	GTEx Portal V6	10
CYP21A2	?	ENSG00000231852	subcutaneous adipocytes	4.22×10^{-8} (p-value)	GTEx Portal V6	25
CYP21A1P	?	ENSG00000204338	subcutaneous adipocytes	1.25×10^{-14} (p-value)	GTEx Portal V6	25
HLA-DQB2	?	ENSG00000232629	visceral adipocytes	4.43×10^{-10} (p-value)	GTEx Portal V6	24
C4A	?	ENSG00000244731	visceral adipocytes	5.89×10^{-11} (p-value)	GTEx Portal V6	24
HLA-C	?	ENSG00000204525	visceral adipocytes	5.84×10^{-7} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	visceral adipocytes	2.87×10^{-9} (p-value)	GTEx Portal V6	24
NOTCH4	?	ENSG00000204301	visceral adipocytes	3.60×10^{-9} (p-value)	GTEx Portal V6	22
C4A	?	ENSG00000244731	stomach	3.95×10^{-16} (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	spleen	3.84×10^{-8} (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	tibial nerve	6.98×10^{-23} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	tibial nerve	6.43×10^{-8} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	tibial nerve	1.34×10^{-13} (p-value)	GTEx Portal V6	25
C4A	?	ENSG00000244731	sigmoid colon	1.97×10^{-9} (p-value)	GTEx Portal V6	24
CYP21A1P	?	ENSG00000204338	sigmoid colon	6.24×10^{-7} (p-value)	GTEx Portal V6	1
LY6G6C	?	ENSG00000204421	esophagus mucosa	4.94×10^{-9} (p-value)	GTEx Portal V6	23
C4A	?	ENSG00000244731	esophagus mucosa	4.61×10^{-16} (p-value)	GTEx Portal V6	25
HLA-C	?	ENSG00000204525	esophagus mucosa	2.15×10^{-9} (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	esophagus mucosa	1.69×10^{-10} (p-value)	GTEx Portal V6	24
MICB	?	ENSG00000204516	esophagus mucosa	9.26×10^{-9} (p-value)	GTEx Portal V6	23
HLA-DQB1	?	ENSG00000179344	esophagus mucosa	1.49×10^{-6} (p-value)	GTEx Portal V6	6
HLA-DMA	?	ENSG00000204257	esophagus mucosa	2.46×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	gastroesophageal junction	1.02×10^{-12} (p-value)	GTEx Portal V6	24
CYP21A1P	?	ENSG00000204338	gastroesophageal junction	1.06×10^{-8} (p-value)	GTEx Portal V6	24
HLA-DQB2	?	ENSG00000232629	gastroesophageal junction	8.86×10^{-7} (p-value)	GTEx Portal V6	5
C4A	?	ENSG00000244731	cerebellar hemisphere	5.31×10^{-8} (p-value)	GTEx Portal V6	22
TNXB	?	ENSG00000168477	adrenal gland	2.46×10^{-7} (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	adrenal gland	5.08×10^{-8} (p-value)	GTEx Portal V6	19
STK19	?	ENSG00000204344	adrenal gland	2.32×10^{-7} (p-value)	GTEx Portal V6	20
STK19B	?	ENSG00000250535	adrenal gland	7.79×10^{-12} (p-value)	GTEx Portal V6	25
TNXA	?	ENSG00000248290	adrenal gland	5.38×10^{-10} (p-value)	GTEx Portal V6	21
CYP21A1P	?	ENSG00000204338	adrenal gland	2.89×10^{-8} (p-value)	GTEx Portal V6	22
C4A	?	ENSG00000244731	testis	3.99×10^{-17} (p-value)	GTEx Portal V6	25

CYP21A1P	?	ENSG00000204338	testis	1.92×10-12 (p-value)	GTEx Portal V6	25
HLA-DMA	?	ENSG00000204257	cerebellum	2.83×10-9 (p-value)	GTEx Portal V6	20
C4A	?	ENSG00000244731	cerebellum	4.80×10-8 (p-value)	GTEx Portal V6	20
CYP21A1P	?	ENSG00000204338	cerebellum	8.19×10-8 (p-value)	GTEx Portal V6	20
C4A	?	ENSG00000244731	frontal cortex	1.94×10-8 (p-value)	GTEx Portal V6	19
NOTCH4	ENST00000474612	ILMN_1711157	adipocyte	1.20×10-9 (p-value)	MuTHER consortium	7
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
HCP5	ENST00000414046	ILMN_1803945	skin	5.66×10-6 (p-value)	MuTHER consortium	6
HCP5	ENST00000541196		adipocyte	3.41×10-6 (p-value)	MuTHER consortium	6
HLA-DPB1	ENST00000418931	ILMN_1749070	skin	4.66×10-9 (p-value)	MuTHER consortium	7
HLA-DPB1	ENST00000416804		blood	3.81×10-10 (p-value)	MuTHER consortium	7
			adipocyte	1.57×10-8 (p-value)	MuTHER consortium	7
MICB	ENST00000538442	ILMN_1708006	skin	3.44×10-9 (p-value)	MuTHER consortium	6
MICB	ENST00000399150					
MICB	ENST00000252229					
PSMB9	ENST00000467593	ILMN_1798233	blood	1.55×10-7 (p-value)	MuTHER consortium	4
PSMB9	ENST00000464863					
VARS2	ENST00000321897	ILMN_1737585	monocyte	2.87×10-8 (p-value)	Fairfax et al.	3
VARS2	ENST00000625423		b-cell	3.50×10-6 (p-value)	Fairfax et al.	3
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
HLA-DPB1	ENST00000418931	ILMN_1749070	monocyte	3.64×10-8 (p-value)	Fairfax et al.	2
HLA-DPB1	ENST00000416804		b-cell	2.63×10-8 (p-value)	Fairfax et al.	2
C4A	?	ENSG00000244731	putamen	2.67×10-8 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	caudate basal ganglia	2.05×10-8 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	hypothalamus	8.80×10-8 (p-value)	GTEx Portal V6	2
C4A	?	ENSG00000244731	coronary artery	8.43×10-7 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	anterior cingulate cortex	1.39×10-8 (p-value)	GTEx Portal V6	1
TAP2	ENST00000620123	ILMN_1759250	blood	8.02×10-7 (p-value)	Westra et al.	1
TAP2	ENST00000374899					
XXbac-BPG246D15.9	ENST00000452392					
TAP2	ENST00000464100					
TAP2	ENST00000374897					
HLA-DQB1	ENST00000399082	209480_at	blood	4.90×10-12 (p-value)	Dixon et al.	1
HLA-DQB1	ENST00000399084					
HLA-DQB1	ENST00000434651					
HLA-DQB1	ENST00000460185					
HLA-DQB1	ENST00000399079					
HLA-DQB1	ENST00000443574					

HLA-DQB1	ENST00000374943					
HLA-DQB1	ENST00000487676					
?	?	221491_x_at	blood	2.80×10^{-17} (p-value)	Dixon et al.	1
HLA-DQA1	ENST00000374949	213831_at	blood	1.60×10^{-10} (p-value)	Dixon et al.	1
HLA-DQA1	ENST00000496318					
HLA-DQA1	ENST00000460633					
HLA-DQA1	ENST00000482745					
HLA-DQA1	ENST00000343139					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445252	1	ENCP00000048214	XXbac-BPG254F23.6 XXbac-BPG254F23.6 XXbac-BPG254F23.6 XXbac-BPG254F23.6 XXbac-BPG254F23.6 XXbac-BPG254F23.6
		ENCP00000048178	EGFL8 EGFL8 EGFL8 EGFL8 EGFL8 EGFL8 EGFL8
		ENCP00000048131	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000048199	HCG23 HCG23 HCG23 HCG23 HCG23 HCG23 HCG23 HCG23
		ENCP00000048206	HLA-DRB6
		ENCP00000048201	BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BTNL2	upstream gene variant	2924	ENST00000374993	NM_019602.1	ENSP00000364132	2
BTNL2	upstream gene variant	2919	ENST00000454136	?	ENSP00000390613	2
BTNL2	upstream gene variant	2924	ENST00000465865	?	ENSP00000420063	2
BTNL2	downstream gene variant, upstream gene variant	2919	ENST00000446536	?	ENSP00000388434	3
BTNL2	upstream gene variant	2924	ENST00000544175	?	ENSP00000443364	2

C6orf10	upstream gene variant	1187	ENST00000533191	NM_001286474.1	ENSP00000431199	3
C6orf10	upstream gene variant	1182	ENST00000617061	?	ENSP00000482001	3
C6orf10	downstream gene variant, upstream gene variant	1222	ENST00000532023	?	ENSP00000432814	4
C6orf10	downstream gene variant, upstream gene variant	1215	ENST00000534588	?	ENSP00000432566	4
C6orf10	upstream gene variant	1187	ENST00000375015	?	ENSP00000364155	3
C6orf10	downstream gene variant, upstream gene variant	1195	ENST00000442822	?	ENSP00000411164	4
C6orf10	upstream gene variant	1222	ENST00000527965	NM_001286475.1	ENSP00000435103	3
C6orf10	upstream gene variant	1215	ENST00000375007	?	ENSP00000364146	3
C6orf10	upstream gene variant	1215	ENST00000447241	NM_006781.4	ENSP00000415517	3
C6orf10	upstream gene variant	1182	ENST00000612031	?	ENSP00000480403	3
HLA-DOB	downstream gene variant	3619	ENST00000488325	?	ENSP00000436618	1
HLA-DOB	downstream gene variant	3903	ENST00000475235	?	?	1
HLA-DOB	downstream gene variant	3619	ENST00000438763	NM_002120.3	ENSP00000390020	1
HLA-DRA	downstream gene variant	1612	ENST00000374982	?	ENSP00000364121	2
HLA-DRA	downstream gene variant	1612	ENST00000395388	NM_019111.4	ENSP00000378786	2
HLA-DRB9	downstream gene variant	2606	ENST00000449413	?	?	2
HNRNPA1P2	upstream gene variant, downstream gene variant	1697	ENST00000432375	?	?	2
RNU6-603P	downstream gene variant	4802	ENST00000411403	?	?	1
XXbac-BPG154L12.4	upstream gene variant	3590	ENST00000425033	?	?	1
XXbac-BPG154L12.5	upstream gene variant	3163	ENST00000611838	?	?	2
XXbac-BPG246D15.9	downstream gene variant	4623	ENST00000452392	?	ENSP00000391806	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BTNL2	ENST00000454136	?	ENSP00000390613	1
BTNL2	ENST00000374993	NM_019602.1	ENSP00000364132	1
BTNL2	ENST00000544175	?	ENSP00000443364	1
BTNL2	ENST00000465865	?	ENSP00000420063	1
C6orf10	ENST00000534588	?	ENSP00000432566	2
C6orf10	ENST00000533191	NM_001286474.1	ENSP00000431199	8
C6orf10	ENST00000375007	?	ENSP00000364146	8
C6orf10	ENST00000527965	NM_001286475.1	ENSP00000435103	8
C6orf10	ENST00000375015	?	ENSP00000364155	8
C6orf10	ENST00000447241	NM_006781.4	ENSP00000415517	8
C6orf10	ENST00000442822	?	ENSP00000411164	8
C6orf10	ENST00000532023	?	ENSP00000432814	2
C6orf10	ENST00000617061	?	ENSP00000482001	8

C6orf10	ENST00000612031	?	ENSP00000480403	8
HLA-DRB9	ENST00000449413	?	?	1
XXbac-BPG154L12.4	ENST00000425033	?	?	3
XXbac-BPG154L12.5	ENST00000611838	?	?	10

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
XXbac-BPG154L12.4	ENST00000425033	?	1