Locus 105 (rs3094005)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,016,550-31,566,168
block size	549,619 bp
variant count	118 variants

Basic features

Conservation/deleteriousness		Linked genes
phyloP	μ = -0.200 [-3.795 – 1.156]	**gene(s) hit or close-by**: AL662801.1, AL671883.1, ATP6V1G2, ATP6V1G2-DDX39B, DDX39B, DDX39B-AS1, DHFRP2, FGFR3P1, HCG22, HCG27, HCP5, HLA-B, HLA-C, HLA-S, LINC01149, LST1, LTA, LTB, MICA, MICB, MIR6891, NCR3, NFKBIL1, PPIAP9, RNU6-283P, SNORD117, SNORD84, TNF, XXbac-BPG16N22.5, XXbac-BPG181B23.6, XXbac-BPG181B23.7, XXbac-BPG248L24.12, XXbac-BPG299F13.14, XXbac-BPG299F13.15, XXbac-BPG299F13.16, Y_RNA, ZDHHC20P2
phastCons	μ = 0.054 [0 – 1]	**eQTL gene(s)**: BTN3A2, C4A, C4B, C6orf10, CCHCR1, CSF3R, CYP21A1P, CYP21A2, DDX39B, FLOT1, HCG22, HCP5, HIST1H2BD, HLA-B, HLA-C, HLA-DPB1, HLA-DQA1, HLA-DQB1, HLA-G, IER3, LINC00243, LST1, LY6G6C, MICA, MICB, NOTCH4, OR5V1, PPP1R18, PSORS1C1, PSORS1C2, RNF5, SKIV2L, SNORD84, STK19, STK19B, TMEM154, TNXA, VARS2, WASF5P, XXbac-BPG248L24.12, XXbac-BPG248L24.13, XXbac-BPG252P9.10
GERP++	μ = -0.098 [-3.15 – 2.1]	**potentially regulated gene(s)**: ABHD16A (×7), BAG6 (×7), C2 (×10), C6orf47 (×8), C6orf48 (×8), CCHCR1 (×8), CFB (×8), CLIC1 (×8), DDR1 (×8), DDX39B (×8), EHMT2 (×8), GPANK1 (×8), GTF2H4 (×8), HLA-B (×8), HSPA1L (×8), LSM2 (×8), LTA (×8), LTB (×8), LY6G5C (×8), LY6G6D (×8), LY6G6E (×8), MICB (×8), MSH5 (×8), NCR3 (×8), NEU1 (×8), POLR2LP (×8), POU5F1 (×8), PRRC2A (×8), PSORS1C2 (×8), SLC44A4 (×8), STK19 (×8), TCF19 (×8), VARS (×8), VARS2 (×8), WASF5P (×8), XXbac-BPG248L24.10 (×8), ZBTB12 (×4), ZDHHC20P2 (×5)
CADD score	μ = 3.433 [0.004 – 16.26]	**disease gene(s)**: C4B, C4A, CYP21A2, VARS2, CSF3R, LTA, NCR3, C2, NFKBIL1, SKIV2L, TNF, CFB, NEU1, HLA-B, HLA-DQB1, HLA-DPB1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Rheumatoid arthritis (ACPA-negative)	$<9.00\times10^{-9}$	GWAS Catalog	24532677	1
Hematology traits	$<5.00\times10^{-7}$	GWAS Catalog	23263863	1
Myasthenia gravis	$<2.00\times10^{-90}$	GWAS Catalog	23055271	1
Metabolic syndrome	$<2.00\times10^{-8}$	GWAS Catalog	22399527	1
Stevens-Johnson syndrome and toxic epidermal necrolysis (SJS-TEN)	$<2.00\times10^{-8}$	GWAS Catalog	21912425	1
Idiopathic membranous nephropathy	$<6.00\times10^{-72}$	GWAS Catalog	21323541	2
Neonatal lupus	$<5.00\times10^{-10}$	GWAS Catalog	20662065	1
Psoriasis	$<1.00\times10^{-9}$	GWAS Catalog	18364390	1
laurylcarnitine	7.88×10^{-5}	Metabolomics GWAS Server	24816252	1
Lupus erythematosus, systemic	1.95×10^{-6}	dbGaP	pha002867	2
Diabetes mellitus, type 1	5.29×10^{-68}	dbGaP	pha002862	1
Breast neoplasms	1.62×10^{-4}	dbGaP	pha002853	1
Lupus erythematosus, systemic	4.00×10^{-15}	dbGaP	pha002848	2

Disease gene annotation

gene	trait	source DB	source entry/link
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
CSF3R	NEUTROPHILIA, HEREDITARY	OMIM	MIM:162830
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
LTA	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988
LTA	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
NCR3	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000

C2	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
	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
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602	OMIM
NFKBIL1	RHEUMATOID ARTHRITIS	OMIM	MIM:180300	OMIM
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
NEU1	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	OMIM
	NEURAMINIDASE DEFICIENCY	OMIM	MIM:256550	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
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	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-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
CSF3R	Chronic neutrophilic leukemia	OrphaNet	OrphaNet:86829	orphanet
	Hereditary neutrophilia	OrphaNet	OrphaNet:279943	

CSF3R	Atypical chronic myeloid leukemia	OrphaNet	OrphaNet:98824	
HLA-DPB1	Chronic berylliosis	OrphaNet	OrphaNet:133	
HLA-DPB1	Granulomatosis with polyangiitis	OrphaNet	OrphaNet:900	
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064	
NEU1	Juvenile sialidosis type 2	OrphaNet	OrphaNet:93399	
NEU1	Sialidosis type 1	OrphaNet	OrphaNet:812	
NEU1	Congenital sialidosis type 2	OrphaNet	OrphaNet:93400	
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
NEU1	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	
	sialidosis (SIALIDOSIS)	DECIPHER	MIM:256550	

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG248L24.13	?	ENSG00000256166	pancreas	1.60×10^{-8} (p-value)	GTEx Portal V6	107
C4A	?	ENSG00000244731	pancreas	3.09×10^{-9} (p-value)	GTEx Portal V6	115
WASF5P	?	ENSG00000231402	pancreas	8.19×10^{-12} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	pancreas	1.80×10^{-8} (p-value)	GTEx Portal V6	110
HLA-B	?	ENSG00000234745	pancreas	3.41×10^{-7} (p-value)	GTEx Portal V6	95
RNF5	?	ENSG00000204308	pancreas	1.58×10^{-6} (p-value)	GTEx Portal V6	31
CYP21A1P	?	ENSG00000204338	pancreas	1.35×10^{-7} (p-value)	GTEx Portal V6	18
C4A	?	ENSG00000244731	muscularis mucosae	4.51×10^{-15} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	muscularis mucosae	2.65×10^{-11} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	muscularis mucosae	2.07×10^{-6} (p-value)	GTEx Portal V6	6
HLA-B	?	ENSG00000234745	muscularis mucosae	2.42×10^{-7} (p-value)	GTEx Portal V6	96
CYP21A1P	?	ENSG00000204338	muscularis mucosae	2.75×10^{-10} (p-value)	GTEx Portal V6	116
MICB	?	ENSG00000204516	muscularis mucosae	2.13×10^{-8} (p-value)	GTEx Portal V6	41
PSORS1C1	?	ENSG00000204540	muscularis mucosae	2.08×10^{-6} (p-value)	GTEx Portal V6	2
C4A	?	ENSG00000244731	lung	1.56×10^{-15} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	lung	5.80×10^{-11} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	lung	7.03×10^{-9} (p-value)	GTEx Portal V6	99
CYP21A1P	?	ENSG00000204338	lung	1.64×10^{-9} (p-value)	GTEx Portal V6	116
MICB	?	ENSG00000204516	lung	1.60×10^{-6} (p-value)	GTEx Portal V6	5
MICA	?	ENSG00000204520	lung	6.23×10^{-6} (p-value)	GTEx Portal V6	1

C4A	?	ENSG00000244731	atrial appendage	5.15×10-15 (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	atrial appendage	5.33×10-11 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	atrial appendage	1.42×10-6 (p-value)	GTEx Portal V6	6
C4B	?	ENSG00000224389	transformed fibroblasts	2.51×10-13 (p-value)	GTEx Portal V6	116
C4A	?	ENSG00000244731	transformed fibroblasts	3.99×10-18 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	transformed fibroblasts	1.59×10-13 (p-value)	GTEx Portal V6	116
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	5.25×10-6 (p-value)	GTEx Portal V6	47
MICB	?	ENSG00000204516	transformed fibroblasts	3.71×10-6 (p-value)	GTEx Portal V6	17
C4A	?	ENSG00000244731	blood	1.08×10-10 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	blood	3.47×10-12 (p-value)	GTEx Portal V6	109
C4B	?	ENSG00000224389	blood	7.08×10-16 (p-value)	GTEx Portal V6	116
CYP21A2	?	ENSG00000231852	blood	5.84×10-7 (p-value)	GTEx Portal V6	53
CCHCR1	?	ENSG00000204536	blood	4.72×10-7 (p-value)	GTEx Portal V6	97
CYP21A1P	?	ENSG00000204338	blood	1.78×10-12 (p-value)	GTEx Portal V6	116
LINC00243	?	ENSG00000214894	blood	1.78×10-11 (p-value)	GTEx Portal V6	102
XXbac-BPG252P9.10	?	ENSG00000272273	blood	5.09×10-6 (p-value)	GTEx Portal V6	2
C4A	?	ENSG00000244731	breast	2.53×10-13 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	breast	5.94×10-12 (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	breast	4.14×10-12 (p-value)	GTEx Portal V6	116
MICB	?	ENSG00000204516	breast	7.43×10-7 (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	tibial artery	1.88×10-16 (p-value)	GTEx Portal V6	116
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	6.92×10-7 (p-value)	GTEx Portal V6	78
C4A	?	ENSG00000244731	tibial artery	4.90×10-8 (p-value)	GTEx Portal V6	39
CYP21A1P	?	ENSG00000204338	tibial artery	1.54×10-7 (p-value)	GTEx Portal V6	15
MICB	?	ENSG00000204516	tibial artery	4.39×10-11 (p-value)	GTEx Portal V6	34
C4A	?	ENSG00000244731	thyroid	7.45×10-12 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	thyroid	1.90×10-10 (p-value)	GTEx Portal V6	115
FLOT1	?	ENSG00000137312	thyroid	4.67×10-8 (p-value)	GTEx Portal V6	98
CCHCR1	?	ENSG00000204536	thyroid	1.10×10-11 (p-value)	GTEx Portal V6	110
PPP1R18	?	ENSG00000146112	thyroid	6.71×10-7 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	thyroid	1.77×10-13 (p-value)	GTEx Portal V6	116
PSORS1C1	?	ENSG00000204540	thyroid	5.44×10-12 (p-value)	GTEx Portal V6	116
PSORS1C2	?	ENSG00000204538	thyroid	4.58×10-8 (p-value)	GTEx Portal V6	31
MICA	?	ENSG00000204520	thyroid	1.10×10-5 (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	skeletal muscle	2.99×10-13 (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	skeletal muscle	8.80×10-21 (p-value)	GTEx Portal V6	116
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	7.46×10-8 (p-value)	GTEx Portal V6	99
CYP21A1P	?	ENSG00000204338	skeletal muscle	7.49×10-7 (p-value)	GTEx Portal V6	51

RNF5	?	ENSG00000204308	skeletal muscle	4.32×10^{-6} (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	skeletal muscle	1.88×10^{-6} (p-value)	GTEx Portal V6	3
PSORS1C1	?	ENSG00000204540	skeletal muscle	3.52×10^{-6} (p-value)	GTEx Portal V6	5
C4A	?	ENSG00000244731	EBV lymphocytes	2.79×10^{-8} (p-value)	GTEx Portal V6	111
HLA-C	?	ENSG00000204525	EBV lymphocytes	1.93×10^{-6} (p-value)	GTEx Portal V6	51
C4A	?	ENSG00000244731	transverse colon	9.56×10^{-11} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	transverse colon	1.03×10^{-9} (p-value)	GTEx Portal V6	111
HLA-C	?	ENSG00000204525	transverse colon	9.57×10^{-10} (p-value)	GTEx Portal V6	116
VARS2	?	ENSG00000137411	transverse colon	2.70×10^{-6} (p-value)	GTEx Portal V6	3
PSORS1C1	?	ENSG00000204540	transverse colon	2.43×10^{-6} (p-value)	GTEx Portal V6	50
C4A	?	ENSG00000244731	liver	3.18×10^{-12} (p-value)	GTEx Portal V6	116
STK19B	?	ENSG00000250535	liver	1.17×10^{-7} (p-value)	GTEx Portal V6	8
CCHCR1	?	ENSG00000204536	liver	1.52×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	sun exposed skin	3.48×10^{-12} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	sun exposed skin	4.28×10^{-13} (p-value)	GTEx Portal V6	116
C4B	?	ENSG00000224389	sun exposed skin	5.52×10^{-10} (p-value)	GTEx Portal V6	116
CYP21A2	?	ENSG00000231852	sun exposed skin	2.48×10^{-9} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	sun exposed skin	2.63×10^{-6} (p-value)	GTEx Portal V6	33
CYP21A1P	?	ENSG00000204338	sun exposed skin	1.00×10^{-8} (p-value)	GTEx Portal V6	115
PSORS1C1	?	ENSG00000204540	sun exposed skin	6.04×10^{-7} (p-value)	GTEx Portal V6	79
VARS2	?	ENSG00000137411	sun exposed skin	7.32×10^{-6} (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	sun exposed skin	7.73×10^{-13} (p-value)	GTEx Portal V6	113
NOTCH4	?	ENSG00000204301	sun exposed skin	1.55×10^{-6} (p-value)	GTEx Portal V6	15
C4A	?	ENSG00000244731	unexposed skin	3.00×10^{-11} (p-value)	GTEx Portal V6	115
HLA-C	?	ENSG00000204525	unexposed skin	6.98×10^{-8} (p-value)	GTEx Portal V6	114
CYP21A1P	?	ENSG00000204338	unexposed skin	1.71×10^{-8} (p-value)	GTEx Portal V6	112
MICB	?	ENSG00000204516	unexposed skin	1.42×10^{-8} (p-value)	GTEx Portal V6	65
CYP21A2	?	ENSG00000231852	unexposed skin	3.27×10^{-6} (p-value)	GTEx Portal V6	1
C4A	?	ENSG00000244731	aorta	9.09×10^{-12} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	aorta	2.76×10^{-10} (p-value)	GTEx Portal V6	116
HLA-B	?	ENSG00000234745	aorta	3.56×10^{-7} (p-value)	GTEx Portal V6	99
PPP1R18	?	ENSG00000146112	aorta	2.23×10^{-7} (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	aorta	6.17×10^{-11} (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	aorta	2.66×10^{-9} (p-value)	GTEx Portal V6	116
C4A	?	ENSG00000244731	left ventricle	2.24×10^{-8} (p-value)	GTEx Portal V6	71
HLA-C	?	ENSG00000204525	left ventricle	5.04×10^{-9} (p-value)	GTEx Portal V6	99
CYP21A1P	?	ENSG00000204338	left ventricle	1.33×10^{-7} (p-value)	GTEx Portal V6	17

PSORS1C1	?	ENSG00000204540	left ventricle	1.37×10^{-8} (p-value)	GTEx Portal V6	105
NOTCH4	?	ENSG00000204301	left ventricle	1.30×10^{-7} (p-value)	GTEx Portal V6	32
C4A	?	ENSG00000244731	subcutaneous adipocytes	3.88×10^{-10} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	1.39×10^{-13} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	subcutaneous adipocytes	1.56×10^{-12} (p-value)	GTEx Portal V6	115
CYP21A1P	?	ENSG00000204338	subcutaneous adipocytes	8.99×10^{-10} (p-value)	GTEx Portal V6	108
VARS2	?	ENSG00000137411	subcutaneous adipocytes	1.76×10^{-6} (p-value)	GTEx Portal V6	3
CYP21A2	?	ENSG00000231852	subcutaneous adipocytes	5.06×10^{-9} (p-value)	GTEx Portal V6	114
PSORS1C1	?	ENSG00000204540	subcutaneous adipocytes	3.60×10^{-7} (p-value)	GTEx Portal V6	57
NOTCH4	?	ENSG00000204301	subcutaneous adipocytes	1.38×10^{-6} (p-value)	GTEx Portal V6	77
MICB	?	ENSG00000204516	subcutaneous adipocytes	4.89×10^{-8} (p-value)	GTEx Portal V6	30
C4A	?	ENSG00000244731	stomach	6.74×10^{-14} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	stomach	9.04×10^{-9} (p-value)	GTEx Portal V6	99
PSORS1C1	?	ENSG00000204540	stomach	1.26×10^{-6} (p-value)	GTEx Portal V6	51
CCHCR1	?	ENSG00000204536	visceral adipocytes	1.20×10^{-8} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	visceral adipocytes	7.22×10^{-11} (p-value)	GTEx Portal V6	116
C4A	?	ENSG00000244731	visceral adipocytes	1.27×10^{-8} (p-value)	GTEx Portal V6	111
NOTCH4	?	ENSG00000204301	visceral adipocytes	9.33×10^{-7} (p-value)	GTEx Portal V6	23
CYP21A1P	?	ENSG00000204338	visceral adipocytes	4.77×10^{-7} (p-value)	GTEx Portal V6	4
C4A	?	ENSG00000244731	tibial nerve	2.20×10^{-16} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	tibial nerve	1.52×10^{-12} (p-value)	GTEx Portal V6	116
CCHCR1	?	ENSG00000204536	tibial nerve	3.84×10^{-10} (p-value)	GTEx Portal V6	109
CYP21A1P	?	ENSG00000204338	tibial nerve	3.79×10^{-13} (p-value)	GTEx Portal V6	116
PSORS1C1	?	ENSG00000204540	tibial nerve	1.86×10^{-7} (p-value)	GTEx Portal V6	69
MICB	?	ENSG00000204516	tibial nerve	8.80×10^{-8} (p-value)	GTEx Portal V6	21
C4A	?	ENSG00000244731	esophagus mucosa	1.33×10^{-14} (p-value)	GTEx Portal V6	116
HLA-C	?	ENSG00000204525	esophagus mucosa	3.24×10^{-13} (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	esophagus mucosa	1.42×10^{-10} (p-value)	GTEx Portal V6	116
MICB	?	ENSG00000204516	esophagus mucosa	2.18×10^{-10} (p-value)	GTEx Portal V6	116
LY6G6C	?	ENSG00000204421	esophagus mucosa	9.70×10^{-10} (p-value)	GTEx Portal V6	112
PSORS1C1	?	ENSG00000204540	esophagus mucosa	6.36×10^{-7} (p-value)	GTEx Portal V6	7
HLA-C	?	ENSG00000204525	gastroesophageal junction	5.10×10^{-7} (p-value)	GTEx Portal V6	18
C4A	?	ENSG00000244731	gastroesophageal junction	2.08×10^{-7} (p-value)	GTEx Portal V6	32
STK19B	?	ENSG00000250535	adrenal gland	2.85×10^{-11} (p-value)	GTEx Portal V6	116
CYP21A1P	?	ENSG00000204338	adrenal gland	6.58×10^{-8} (p-value)	GTEx Portal V6	110
PSORS1C1	?	ENSG00000204540	adrenal gland	3.76×10^{-7} (p-value)	GTEx Portal V6	82
STK19	?	ENSG00000204344	adrenal gland	7.61×10^{-7} (p-value)	GTEx Portal V6	32

HLA-C	?	ENSG00000204525	adrenal gland	1.04×10-6 (p-value)	GTEx Portal V6	42
TNXA	?	ENSG00000248290	adrenal gland	2.42×10-7 (p-value)	GTEx Portal V6	35
C4A	?	ENSG00000244731	adrenal gland	1.49×10-7 (p-value)	GTEx Portal V6	20
C4A	?	ENSG00000244731	cerebellar hemisphere	8.98×10-10 (p-value)	GTEx Portal V6	49
MICB	?	ENSG00000204516	cerebellar hemisphere	8.82×10-8 (p-value)	GTEx Portal V6	71
C4A	?	ENSG00000244731	testis	1.75×10-11 (p-value)	GTEx Portal V6	110
HLA-C	?	ENSG00000204525	testis	6.97×10-7 (p-value)	GTEx Portal V6	48
CYP21A1P	?	ENSG00000204338	testis	2.49×10-9 (p-value)	GTEx Portal V6	100
C6orf10	?	ENSG00000204296	testis	2.15×10-6 (p-value)	GTEx Portal V6	16
VARS2	ENST00000321897	ILMN_1737585	skin	6.13×10-8 (p-value)	MuTHER consortium	22
VARS2	ENST00000625423		adipocyte	3.43×10-6 (p-value)	MuTHER consortium	22
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
HCG22	ENST00000570223	ILMN_1667229	blood	1.28×10-7 (p-value)	MuTHER consortium	22
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
HCP5	ENST00000414046	ILMN_1803945	skin	2.86×10-9 (p-value)	MuTHER consortium	22
HCP5	ENST00000541196		adipocyte	1.34×10-7 (p-value)	MuTHER consortium	22
MICB	ENST00000538442	ILMN_1708006	skin	7.98×10-14 (p-value)	MuTHER consortium	22
MICB	ENST00000399150					
MICB	ENST00000252229					
NOTCH4	ENST00000474612	ILMN_1711157	adipocyte	9.08×10-8 (p-value)	MuTHER consortium	21
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
LST1	ENST00000339530	ILMN_1718936	blood	3.68×10-5 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
SKIV2L	ENST00000484835	ILMN_1666512	skin	2.93×10-5 (p-value)	MuTHER consortium	1
SKIV2L	ENST00000375394					
SKIV2L	ENST00000474839					
SKIV2L	ENST00000470453					
SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					

SKIV2L	ENST00000485349					
?	?	ILMN_1679389	b-cell	1.55×10-8 (p-value)	Fairfax et al.	9
VARS2	ENST00000321897	ILMN_1737585	monocyte	7.69×10-8 (p-value)	Fairfax et al.	9
VARS2	ENST00000625423		b-cell	8.53×10-6 (p-value)	Fairfax et al.	9
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
OR5V1	ENST00000543825	ILMN_1683352	b-cell	8.01×10-7 (p-value)	Fairfax et al.	9
OR5V1	ENST00000377154					
IER3	ENST00000376377	ILMN_1682717	monocyte	1.79×10-5 (p-value)	Fairfax et al.	8
IER3	ENST00000259874					
DDX39B	ENST00000481456	ILMN_3245672	b-cell	6.77×10-4 (p-value)	Fairfax et al.	3
SNORD84	ENST00000584275					
HLA-C	ENST00000470363	216526_x_at	blood	9.00×10-9 (p-value)	Dixon et al.	1
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000466892					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	?	ENSG00000204525	cerebellum	2.07×10-6 (p-value)	GTEx Portal V6	2
CYP21A1P	?	ENSG00000204338	cerebellum	5.38×10-7 (p-value)	GTEx Portal V6	49
MICB	?	ENSG00000204516	cerebellum	4.33×10-11 (p-value)	GTEx Portal V6	110
C4A	?	ENSG00000244731	cerebellum	2.67×10-7 (p-value)	GTEx Portal V6	3
C4A	?	ENSG00000244731	frontal cortex	4.09×10-9 (p-value)	GTEx Portal V6	46
CCHCR1	?	ENSG00000204536	sigmoid colon	1.95×10-6 (p-value)	GTEx Portal V6	4
C4A	?	ENSG00000244731	sigmoid colon	3.19×10-7 (p-value)	GTEx Portal V6	9
?	?	ILMN_1883997	blood	1.67×10-5 (p-value)	Westra et al.	1
HCP5	ENST00000414046	ILMN_1803945	blood	3.07×10-6 (p-value)	Westra et al.	2
HCP5	ENST00000541196					
?	?	ILMN_1893395	monocyte	6.84×10-28 (p-value)	Zeller et al.	2
VARS2	ENST00000321897	ILMN_1737585	monocyte	1.60×10-12 (p-value)	Zeller et al.	1
VARS2	ENST00000625423					
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
?	?	ILMN_1666078	monocyte	4.19×10-19 (p-value)	Zeller et al.	2
HLA-C	ENST00000470363	ILMN_2150787	monocyte	9.48×10-30 (p-value)	Zeller et al.	2
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					

HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						
?	?	ILMN_1671054	monocyte		1.03×10^{-30} (p-value)	Zeller et al.	2
HLA-G	ENST00000376828	ILMN_1656670	monocyte		2.47×10^{-13} (p-value)	Zeller et al.	1
HLA-G	ENST00000360323						
HLA-G	ENST00000478519						
HLA-G	ENST00000478355						
HLA-G	ENST00000428701						
HLA-G	ENST00000376815						
HLA-G	ENST00000376818						
BTN3A2	ENST00000396934	ILMN_1676528	monocyte		2.50×10^{-21} (p-value)	Zeller et al.	2
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
?	?	ILMN_1883997	monocyte		8.89×10^{-89} (p-value)	Zeller et al.	2
CYP21A1P	?	ENSG00000204338	caudate basal ganglia		4.92×10^{-7} (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
HLA-DQB1	ENST00000399082	209480_at	chr6	blood	4.30×10^{-11} (p-value)	Dixon et al.	3
HLA-DQB1	ENST00000399084						
HLA-DQB1	ENST00000434651						
HLA-DQB1	ENST00000460185						
HLA-DQB1	ENST00000399079						
HLA-DQB1	ENST00000443574						
HLA-DQB1	ENST00000374943						
HLA-DQB1	ENST00000487676						
?	?	221491_x_at	chr6	blood	1.20×10^{-14} (p-value)	Dixon et al.	3
HLA-DQA1	ENST00000374949	213831_at	chr6	blood	2.40×10^{-9} (p-value)	Dixon et al.	3
HLA-DQA1	ENST00000496318						
HLA-DQA1	ENST00000460633						
HLA-DQA1	ENST00000482745						
HLA-DQA1	ENST00000343139						
CSF3R	ENST00000464465	ILMN_2371280	chr1	blood	1.51×10^{-8} (p-value)	Westra et al.	1
CSF3R	ENST00000373104						
CSF3R	ENST00000487540						
CSF3R	ENST00000373106						
CSF3R	ENST00000331941						
CSF3R	ENST00000484762						
CSF3R	ENST00000480825						
CSF3R	ENST00000361632						
CSF3R	ENST00000373103						
HIST1H2BD	ENST00000289316	ILMN_1651496	chr6	blood	5.26×10^{-10} (p-value)	Westra et al.	2
TMEM154	ENST00000304385	ILMN_2088124	chr4	blood	7.64×10^{-24} (p-value)	Westra et al.	2
BTN3A2	ENST00000396934	ILMN_1676528	chr6	blood	1.39×10^{-10} (p-value)	Westra et al.	1
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
?	?	ILMN_1752592	chr6	monocyte	3.44×10^{-55} (p-value)	Zeller et al.	2
?	?	ILMN_1715169	chr6	monocyte	1.81×10^{-19} (p-value)	Zeller et al.	2
HLA-DPB1	ENST00000418931	ILMN_1749070	chr6	monocyte	6.72×10^{-18} (p-value)	Zeller et al.	2

HLA-DPB1	ENST00000416804						
?	?	ILMN_1661266	chr6	monocyte	2.70×10-25 (p-value)	Zeller et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444609	2	ENCP00000047951	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
		ENCP00000048082	LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D LY6G6D LY6G6E LY6G6D LY6G6D LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D
		ENCP00000047987	POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1
ENCE00000444610	3	ENCP00000047951	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
		ENCP00000048071	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000047980	PSORS1C2 PSORS1C2 POLR2LP PSORS1C2 POLR2LP PSORS1C2 PSORS1C2 PSORS1C2 POLR2LP POLR2LP POLR2LP POLR2LP
		ENCP00000047987	POU5F1 POU5F1 POU5F1 POU5F1 POU5F1

			POU5F1
			POU5F1
		ENCP00000047988	POU5F1
			POU5F1
			POU5F1
			POU5F1
			POU5F1
			POU5F1
			POU5F1
		ENCP00000047969	VARS2
			VARS2
			VARS2
			VARS2
			VARS2
			VARS2
ENCE00000445031	2	ENCP00000048032	LTA
			LTA
			LTA
			LTA
			LTA
			LTA
			LTA
		ENCP00000048026	DDX39B
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			DDX39B
ENCE00000444689	1	ENCP00000048034	LTB
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			LTB
			LTB
			LTB
			LTB
			LTB
			LTB
		ENCP00000048002	XXbac-BPG248L24.10
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			XXbac-BPG248L24.10
			XXbac-BPG248L24.10
			XXbac-BPG248L24.10
		ENCP00000048058	BAG6
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		ENCP00000048114	HSPA1L
			HSPA1L
			HSPA1L
			HSPA1L
			HSPA1L
		ENCP00000048038	NCR3
			NCR3
			NCR3
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			NCR3
			NCR3
			NCR3
			NCR3
		ENCP00000048003	HLA-B
			HLA-B
			HLA-B
			HLA-B
			HLA-B

Locus 105 (sentinel SNP rs3094005 + 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.

			HLA-B
		ENCP00000048004	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048039	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
		ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048032	LTA LTA LTA LTA LTA LTA LTA
		ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048026	DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B
		ENCP00000047963	GTF2H4 GTF2H4 GTF2H4 GTF2H4 GTF2H4 GTF2H4
		ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
		ENCP00000048078	ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A
ENCE00000445159	1	ENCP00000048082	LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D

			LY6G6D LY6G6E LY6G6D LY6G6D LY6G6E LY6G6E LY6G6E LY6G6D LY6G6D
ENCE00000444953	1	ENCP00000048010	ZDHHC20P2 ZDHHC20P2 ZDHHC20P2 ZDHHC20P2 ZDHHC20P2
		ENCP00000048049	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCE00000444759	1	ENCP00000048071	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048129	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000047980	PSORS1C2 PSORS1C2 POLR2LP PSORS1C2 POLR2LP PSORS1C2 PSORS1C2 PSORS1C2 POLR2LP POLR2LP POLR2LP POLR2LP
		ENCP00000047987	POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1
		ENCP00000047988	POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1 POU5F1
ENCE00000444783	1	ENCP00000047982	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
		ENCP00000048000	WASF5P WASF5P

			WASF5P WASF5P WASF5P WASF5P WASF5P
		ENCP00000047981	CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1 CCHCR1
ENCE00000444795	1	ENCP00000048115	C6orf48 C6orf48 C6orf48 C6orf48 C6orf48
		ENCP00000048132	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000048076	ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A ABHD16A
		ENCP00000048055	BAG6 BAG6 BAG6 BAG6 BAG6 BAG6 BAG6
		ENCP00000048135	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000048105	VARS VARS VARS VARS VARS VARS VARS
		ENCP00000048130	ZBTB12 ZBTB12 ZBTB12 ZBTB12 ZBTB12
		ENCP00000048102	VARS VARS VARS VARS VARS VARS VARS
		ENCP00000048091	CLIC1 CLIC1 CLIC1 CLIC1 CLIC1

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ENCP00000048123	EHMT2
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	EHMT2
ENCP00000048092	CLIC1
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	CLIC1
	CLIC1
	MSH5
	CLIC1
	CLIC1
	CLIC1
	MSH5
	MSH5
	MSH5
	MSH5
	MSH5
	MSH5
	MSH5
ENCP00000048103	VARS
	VARS
	VARS
	VARS
	VARS
	VARS
	VARS
ENCP00000048070	LY6G5C
	LY6G5C
	LY6G5C
	LY6G5C
	LY6G5C
	LY6G5C
	LY6G5C
ENCP00000048054	BAG6
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ENCP00000048140	CFB
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	CFB
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	CFB
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ENCP00000048077	ABHD16A
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ENCP00000048090	CLIC1
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ENCP00000047984	TCF19
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	ENCP00000048061	C6orf47
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	ENCP00000048119	SLC44A4
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		NEU1
	ENCP00000048111	LSM2
		LSM2
		LSM2
		LSM2
		LSM2
		LSM2
	ENCP00000048127	EHMT2
		EHMT2
		EHMT2
		EHMT2
		EHMT2
		EHMT2
		EHMT2
	ENCP00000048154	STK19
		STK19
		STK19
		STK19
		STK19
		STK19
	ENCP00000048128	C2
		C2
		EHMT2
		EHMT2
		C2
		EHMT2
		C2
		EHMT2
		EHMT2
		C2
		C2
		C2
		EHMT2
		C2
		EHMT2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487811 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	DNase1, H3K4me2
		blood (K562)	Ini1, H3K27ac, Max, H3K9ac, H2AZ, H3K4me3, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		liver (HepG2)	DNase1, FOSL2, Jund, Srf, H3K4me3, PolII, H3K4me1, H3K4me2, FOXA1, H3K27ac, H3K36me3

		blood (GM12878)	IRF4, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1, H2AZ, Cjun, PolII, Srf, BATF
		lung (IMR90)	H3K4me3, H3K4me2, DNase1, H4K8ac
		nervous (NH-A)	DNase1, H3K4me2
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me2
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3, H3K9ac, DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K36me3, PolII, H3K4me2, Cjun
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001703036 (CTCF binding site)	3	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	CTCF
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001404918 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3, H3K36me3
		blood (GM12878)	POU2F2, PolII, H3K4me3, H3K4me2, H3K36me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K36me3, H3K27me3
ENSR00001703038 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, Yy1, H3K4me2, DNase1
		HSMMtube	H3K27me3, CTCF, DNase1
		blood (K562)	H3K27me3, PolII, H2AZ, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF, DNase1, H3K27me3
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	Rad21, H2AZ, H3K4me2, CTCF, H3K27me3, DNase1
		lung (IMR90)	DNase1, H3K27me3, CTCF
		blood (GM12878)	DNase1, H3K4me3, H2AZ, Rad21, H3K4me2, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	H3K4me2, CTCF, H3K27me3
		NHLF	CTCF, H3K27me3
		Osteobl	H3K4me2, CTCF, H3K27me3
		blood (DND-41)	H3K4me1, CTCF, H3K27me3
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF, H3K27me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me3, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3, CTCF
		A549	H3K27me3, DNase1, CTCF, H3K4me3
ENSR00001703043 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00000662334 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, PolII, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF, H3K4me3, H3K4me2
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me2, CTCF, H3K27me3, DNase1
		lung (IMR90)	H3K27me3, CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	CTCF, DNase1
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3, CTCF
		A549	CTCF

ENSR00001703045 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000129758 (promoter flanking region)	4	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, DNase1, TAF7, PolII, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H2AZ, H3K36me3
		blood (K562)	H3K27me3, Cfos
		skin (NHDF-AD)	DNase1, H3K27me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		A549	DNase1, H3K4me3
		skin (NHEK)	H3K4me3, DNase1, H3K4me2, H3K9ac
ENSR00000264406 (promoter)	1	embryonic stem cell (H1ESC)	H3K9ac, H3K36me3, H3K27me3, PolII, Rad21, TAF7, CTCF, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	CTCF, H3K4me2, DNase1, H3K27me3, H2AZ
		blood (K562)	Max, ZBTB7A, Rad21, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Ini1, H3K27me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K4me2
		liver (HepG2)	DNase1, H3K27me3, PolII, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3
		lung (IMR90)	H3K4me2, H3K4me3, CTCF
		blood (GM12878)	PolII, H2AZ, DNase1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	H3K9ac, H4K20me1, H3K4me3, H3K27ac, H3K4me2, DNase1, H3K4me1, CTCF
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000487848 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, PolII, TAF7, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	Max, ZBTB7A, CTCF, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2
		liver (HepG2)	PolII, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4me2
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K4me2, H3K4me3, DNase1, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, CTCF, H3K4me1
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K4me3
		endothelium (HUVEC)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac
ENSR00000487857 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, Egr1, FOSL1, Gabp, Sin3Ak20, CTCF, SIX5, Yy1, TAF1, H3K4me2, SP1, H3K4me3, H3K36me3, H3K27me3, PolII
		HSMMtube	DNase1, H3K36me3, H3K4me2, H3K9ac, CTCF
		blood (K562)	H2AZ, Rad21, TAF1, ELF1, PolII, H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, H3K79me2, Gabp, H3K27me3, Brg1, SIX5, H3K27ac, Max, H3K4me1, H3K9ac
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H3K9ac, H3K36me3, DNase1, CTCF, H3K4me2
		liver (HepG2)	Gabp, PolII, Rad21, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc,

		blood (GM12878)	H3K4me3, H3K36me3, DNase1 H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac, H3K79me2, Rad21, SIX5, PolII, BCLAF1, H2AZ, ETS1, DNase1, Yy1, Gabp, ELF1
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3, CTCF
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2, H3K36me3
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1
		NHLF	DNase1, CTCF, H3K27ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me3, H3K36me3, H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac
		breast (HMEC)	DNase1, H3K4me3, H3K4me1, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, H3K4me3, Gabp, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	CTCF, H3K4me2, H3K36me3, H3K4me3, DNase1
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, PolII, H3K4me2, H3K9ac
		A549	H3K36me3, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00001703058 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Egr1, FOSL1, Gabp, Sin3Ak20, CTCF, Yy1, TAF1, H3K4me2, SP1, H3K4me3
		HSMMtube	CTCF, H3K4me2, DNase1
		blood (K562)	H3K27me3, PolII, Brg1, DNase1, H3K27ac, H2AZ, Rad21, H3K4me3, H3K4me2, CTCF
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	CTCF, H3K4me2, DNase1
		liver (HepG2)	Rad21, H2AZ, H3K4me2, CTCF, H3K4me3, DNase1, Gabp
		lung (IMR90)	CTCF, H3K4me3, H3K4me2, DNase1
		blood (GM12878)	H2AZ, DNase1, Rad21, H3K4me3, H3K4me2, CTCF
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	DNase1, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K4me3, Gabp, CTCF
		monocytes (Monocytes-CD14+)	H3K4me3, DNase1, CTCF, H3K4me2
		endothelium (HUVEC)	H3K36me3, DNase1, H3K4me2, H3K9ac, CTCF
		A549	H3K4me3, H3K4me2, DNase1, CTCF, H3K9ac
ENSR00000487869 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, Nrsf
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K36me3
		skin (NHEK)	DNase1
ENSR00000264421 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, Yy1, DNase1
		HSMMtube	DNase1, CTCF
		blood (K562)	PolII, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H2AZ, H3K4me2, CTCF, DNase1
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF, Yy1, H2AZ, DNase1
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF, DNase1
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	DNase1, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL662801.1	upstream gene variant	4834	ENST00000617927	?	?	1
AL671883.1	downstream gene variant,	2210	ENST00000578513	?	?	3

ATP6V1G2-DDX39B	upstream gene variant downstream gene variant	2711	ENST00000480131 ?	ENSP00000420191	1
ATP6V1G2-DDX39B	downstream gene variant	4495	ENST00000475917 ?	?	1
DDX39B	upstream gene variant	4481	ENST00000428450 ?	ENSP00000405707	1
DDX39B	upstream gene variant	4453	ENST00000462256 ?	?	1
DDX39B	upstream gene variant	4452	ENST00000428098 ?	ENSP00000392672	1
DDX39B	upstream gene variant	2228	ENST00000484566 ?	ENSP00000436220	1
DDX39B	upstream gene variant	4939	ENST00000427214 ?	ENSP00000399371	1
DDX39B	upstream gene variant	4069	ENST00000482195 ?	?	1
DDX39B	upstream gene variant	4986	ENST00000419338 ?	ENSP00000410313	1
DDX39B	upstream gene variant	4469	ENST00000481456 ?	?	1
DDX39B	upstream gene variant	4489	ENST00000431908 ?	ENSP00000408000	1
DDX39B	upstream gene variant	4741	ENST00000449757 ?	ENSP00000409426	1
DDX39B	upstream gene variant	3192	ENST00000417023 ?	ENSP00000406422	1
DDX39B	downstream gene variant, upstream gene variant	1116	ENST00000456976 ?	ENSP00000393984	2
DDX39B	downstream gene variant, upstream gene variant	1128	ENST00000418897 ?	ENSP00000399841	2
DDX39B	upstream gene variant	4022	ENST00000396172 NM_004640.6	ENSP00000379475	1
DDX39B	upstream gene variant	4022	ENST00000458640 NM_080598.5	ENSP00000416269	1
DDX39B	downstream gene variant, upstream gene variant	1179	ENST00000419020 ?	ENSP00000405245	2
DDX39B	upstream gene variant	4533	ENST00000456662 ?	ENSP00000416350	1
DDX39B-AS1	upstream gene variant, downstream gene variant	3343	ENST00000420520 ?	?	2
DDX39B-AS1	upstream gene variant, downstream gene variant	3332	ENST00000416684 ?	?	2
DHFRP2	downstream gene variant, upstream gene variant	756	ENST00000414224 ?	?	8
FGFR3P1	upstream gene variant, downstream gene variant	806	ENST00000449999 ?	?	4
HCG22	upstream gene variant	3076	ENST00000426185 ?	?	1
HCG22	upstream gene variant	2319	ENST00000565192 ?	?	2
HCG22	upstream gene variant	2369	ENST00000562344 ?	?	2
HCG22	upstream gene variant	3071	ENST00000570223 ?	?	1
HCG22	upstream gene variant	3076	ENST00000566475 ?	?	1
HCG22	upstream gene variant	2941	ENST00000615046 ?	?	1
HCG27	downstream gene variant	406	ENST00000383331 ?	ENSP00000453364	1
HCG27	downstream gene variant	1152	ENST00000424675 ?	ENSP00000453627	1
HCG27	downstream gene variant	3890	ENST00000414008 ?	?	1
HCG27	downstream gene variant	1373	ENST00000415276 ?	?	1
HCP5	upstream gene variant, downstream gene variant	215	ENST00000460889 ?	?	10
HCP5	upstream gene variant, downstream gene variant	268	ENST00000467369 ?	?	8
HCP5	upstream gene variant, downstream gene variant	212	ENST00000541196 ?	?	10
HCP5	downstream gene variant	107	ENST00000414046 ?	?	6

HLA-B	downstream gene variant, upstream gene variant	382	ENST00000412585	NM_005514.6	ENSP00000399168	14
HLA-B	downstream gene variant, upstream gene variant	1109	ENST00000434333	?	ENSP00000405931	10
HLA-B	downstream gene variant, upstream gene variant	1632	ENST00000463574	?	?	6
HLA-B	downstream gene variant, upstream gene variant	382	ENST00000497377	?	?	9
HLA-B	downstream gene variant, upstream gene variant	1392	ENST00000474381	?	?	9
HLA-B	downstream gene variant, upstream gene variant	382	ENST00000481849	?	?	9
HLA-B	downstream gene variant, upstream gene variant	1118	ENST00000498007	?	?	10
HLA-C	upstream gene variant	1139	ENST00000620806	?	ENSP00000480331	7
HLA-S	downstream gene variant, upstream gene variant	1332	ENST00000425174	?	?	6
LINC01149	upstream gene variant	1115	ENST00000430364	?	?	1
LST1	downstream gene variant	2115	ENST00000211921	?	ENSP00000211921	1
LST1	downstream gene variant	2263	ENST00000376092	?	ENSP00000365260	1
LST1	downstream gene variant	2016	ENST00000396101	?	ENSP00000379408	1
LST1	downstream gene variant	2016	ENST00000303757	NM_205840.2	ENSP00000303649	1
LST1	downstream gene variant	2016	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	downstream gene variant	2016	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	downstream gene variant	2016	ENST00000376096	?	ENSP00000365264	1
LST1	downstream gene variant	2016	ENST00000376090	?	ENSP00000365258	1
LST1	downstream gene variant	2115	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	downstream gene variant	2168	ENST00000376110	?	ENSP00000365278	1
LST1	downstream gene variant	2168	ENST00000460834	?	?	1
LST1	downstream gene variant	2016	ENST00000464526	?	?	1
LST1	downstream gene variant	2249	ENST00000490742	?	ENSP00000418578	1
LST1	downstream gene variant	3084	ENST00000419073	?	?	1
LST1	downstream gene variant	3195	ENST00000433492	?	ENSP00000399869	1
LST1	downstream gene variant	2016	ENST00000376089	?	ENSP00000365257	1
LST1	downstream gene variant	2016	ENST00000376099	?	ENSP00000365267	1
LST1	downstream gene variant	2016	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	downstream gene variant	2017	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	downstream gene variant	2168	ENST00000464044	?	ENSP00000417645	1
LST1	downstream gene variant	2299	ENST00000396112	?	ENSP00000379418	1
LTA	downstream gene variant	4749	ENST00000489638	?	?	1
LTA	downstream gene variant	4749	ENST00000471842	?	?	1
LTA	downstream gene variant	4749	ENST00000454783	NM_001159740.2	ENSP00000403495	1
LTA	downstream gene variant	4749	ENST00000418386	NM_000595.3	ENSP00000413450	1
LTB	downstream gene variant	1482	ENST00000446745	NM_009588.1	ENSP00000416113	1

LTB	downstream gene variant	1551	ENST00000482429	?	?	1
LTB	downstream gene variant	1452	ENST00000483972	?	?	1
LTB	downstream gene variant	1482	ENST00000429299	NM_002341.1	ENSP00000410481	1
MICA	downstream gene variant	3039	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	downstream gene variant	3039	ENST00000421350	?	ENSP00000402410	1
MICA	downstream gene variant	3041	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICB	upstream gene variant, downstream gene variant	523	ENST00000538442	NM_001289160.1	ENSP00000442345	3
MICB	upstream gene variant, downstream gene variant	845	ENST00000252229	NM_005931.4	ENSP00000252229	4
MICB	upstream gene variant, downstream gene variant	816	ENST00000399150	NM_001289161.1	ENSP00000382103	4
MICB	upstream gene variant	3938	ENST00000494577	?	?	1
MIR6891	downstream gene variant, upstream gene variant	1734	ENST00000618788	?	?	6
NFKBIL1	upstream gene variant	1191	ENST00000473655	?	?	1
NFKBIL1	upstream gene variant	400	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	1
NFKBIL1	upstream gene variant	1143	ENST00000376145	NM_001144961.1	ENSP00000365315	1
NFKBIL1	upstream gene variant	1170	ENST00000496233	?	ENSP00000437148	1
NFKBIL1	upstream gene variant	1111	ENST00000376148	NM_005007.3	ENSP00000365318	1
PPIAP9	downstream gene variant	3776	ENST00000403866	?	?	1
RNU6-283P	upstream gene variant, downstream gene variant	827	ENST00000364788	?	?	6
SNORD117	upstream gene variant	1254	ENST00000364915	?	?	1
SNORD84	downstream gene variant	3398	ENST00000584275	?	?	1
TNF	downstream gene variant	737	ENST00000449264	NM_000594.3	ENSP00000398698	1
XXbac-BPG16N22.5	upstream gene variant	275	ENST00000538358	?	?	2
XXbac-BPG181B23.6	downstream gene variant, upstream gene variant	440	ENST00000440087	?	?	9
XXbac-BPG181B23.7	downstream gene variant	1403	ENST00000606743	?	?	1
XXbac-BPG248L24.12	upstream gene variant, downstream gene variant	660	ENST00000603274	?	?	8
XXbac-BPG299F13.14	upstream gene variant	2456	ENST00000606909	?	?	1
XXbac-BPG299F13.15	downstream gene variant	3713	ENST00000546340	?	?	1
XXbac-BPG299F13.16	upstream gene variant	1748	ENST00000538455	?	?	1
Y_RNA	downstream gene variant, upstream gene variant	480	ENST00000383850	?	?	2
ZDHHC20P2	upstream gene variant, downstream gene variant	1535	ENST00000424108	?	?	5

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V1G2	ENST00000376151	NM_001204078.1	ENSP00000365321	1
ATP6V1G2	ENST00000303892	NM_130463.3	ENSP00000302194	1
ATP6V1G2	ENST00000481998	?	?	1

ATP6V1G2-DDX39B	ENST00000376185	?	ENSP00000365356	1
ATP6V1G2-DDX39B	ENST00000475917	?	?	1
ATP6V1G2-DDX39B	ENST00000480131	?	ENSP00000420191	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V1G2	ENST00000415099	?	ENSP00000390148	1
ATP6V1G2	ENST00000483170	?	?	1
ATP6V1G2	ENST00000483251	NM_138282.2	ENSP00000419698	1
ATP6V1G2-DDX39B	ENST00000376185	?	ENSP00000365356	2
DDX39B	ENST00000427214	?	ENSP00000399371	1
DDX39B	ENST00000376177	?	ENSP00000365347	1
DDX39B	ENST00000449757	?	ENSP00000409426	1
DDX39B	ENST00000456662	?	ENSP00000416350	1
DDX39B	ENST00000462256	?	?	1
DDX39B	ENST00000458640	NM_080598.5	ENSP00000416269	1
DDX39B	ENST00000396172	NM_004640.6	ENSP00000379475	1
DDX39B	ENST00000481456	?	?	1
DDX39B	ENST00000419338	?	ENSP00000410313	1
DDX39B	ENST00000431908	?	ENSP00000408000	1
DDX39B	ENST00000482195	?	?	1
DDX39B	ENST00000428098	?	ENSP00000392672	1
DDX39B	ENST00000428450	?	ENSP00000405707	1
HCP5	ENST00000467369	?	?	7
HCP5	ENST00000414046	?	?	10
HLA-C	ENST00000620806	?	ENSP00000480331	57
LINC01149	ENST00000430364	?	?	2
MICA	ENST00000449934	NM_001177519.2	ENSP00000413079	1
MICA	ENST00000421350	?	ENSP00000402410	1
MICA	ENST00000616296	NM_001289152.1, NM_001289153.1, NM_001289154.1	ENSP00000482382	1
MICB	ENST00000252229	NM_005931.4	ENSP00000252229	2
MICB	ENST00000538442	NM_001289160.1	ENSP00000442345	3
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	2
NCR3	ENST00000491161	?	?	1
NCR3	ENST00000376071	?	ENSP00000365239	1
NCR3	ENST00000376072	NM_001145467.1	ENSP00000365240	1
NCR3	ENST00000376073	NM_001145466.1	ENSP00000365241	1
NCR3	ENST00000340027	NM_147130.2	ENSP00000342156	1
NCR3	ENST00000495600	?	?	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
DHFRP2	ENST00000414224	?	1

HCP5	ENST00000414046	?	3
HCP5	ENST00000541196	?	1
LINC01149	ENST00000430364	?	1
MICB	ENST00000494577	?	1
XXbac-BPG181B23.6	ENST00000440087	?	1
ZDHHC20P2	ENST00000424108	?	2