Locus 180 (rs3132645)

SNiPAcard

Block annotations

Block info

genomic range	chr6:29,970,960-30,792,117
block size	821,158 bp
variant count	107 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.141 [-3.802 – 4.141]	**gene(s) hit or close-by**	ABCF1, AL662797.1, AL662800.2, ATAT1, C6orf136, DHX16, FLOT1, GNL1, HCG17, HCG18, HCG19P, HCG20, HCG4P3, HLA-J, HLA-L, HLA-N, IER3, LINC00243, MDC1, MICC, MRPS18B, NRM, PAIP1P1, PPP1R10, PPP1R11, PPP1R18, PTMAP1, RN7SL353P, RNF39, RPP21, SUCLA2P1, TRIM10, TRIM15, TRIM26, TRIM26BP, TRIM31, TRIM31-AS1, TRIM39, TRIM39-RPP21, TRIM40, TUBB, UBQLN1P1, XXbac-BPG252P9.10, XXbac-BPG252P9.9, XXbac-BPG283O16.9, Y_RNA, ZNRD1, ZNRD1-AS1, ZNRD1-AS1_3
phastCons	μ = 0.130 [0 – 1]	**eQTL gene(s)**	BTN3A2, BTN3A3, C2, CCHCR1, CFB, FLOT1, GABBR1, HCG17, HCG20, HCG22, HCP5, HIST1H3I, HLA-A, HLA-B, HLA-C, HLA-F-AS1, HLA-G, HLA-H, HLA-J, HLA-K, HLA-L, HLA-U, HLA-V, IER3, LINC00243, MICB, NDUFS1, OR5V1, PPP1R18, PSORS1C1, RNF39, RPP21, TMEM154, TRIM26BP, TRIM31, TRIM39-RPP21, VARS2, WASF5P, ZFP57, ZNRD1, ZNRD1-AS1_3
GERP++	μ = -0.462 [-9.98 – 5.1]	**potentially regulated gene(s)**	ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, ATAT1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, FLOT1, HCG19P, HCG19P, HCG19P, HCG19P, HCG19P, HCG19P, HCG19P, NRM, NRM, NRM, NRM, NRM, NRM, NRM, RNF39, RNF39, RNF39, RNF39, RNF39, RNF39, RNF39, RNF39, TRIM26, TRIM26, TRIM26, TRIM26, TRIM26, TRIM26, TRIM26, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31, TRIM31
CADD score	μ = 4.127 [0.015 – 22.4]	**disease gene(s)**	ZFP57, NDUFS1, C2, CFB, VARS2, TUBB, HLA-A, HLA-B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Autism spectrum disorder, attention deficit-hyperactivity disorder, bipolar disorder, major depressive disorder, and schizophrenia (combined)	$<8.00\times10^{-9}$	GWAS Catalog	23453885	1
AIDS progression	$<5.00\times10^{-7}$	GWAS Catalog	19115949	1
3-indoxyl sulfate	8.34×10^{-5}	Metabolomics GWAS Server	24816252	1
Lupus erythematosus, systemic	6.18×10^{-6}	dbGaP	pha002867	3
Lupus erythematosus, systemic	6.09×10^{-10}	dbGaP	pha002848	3

Disease gene annotation

gene	trait	source DB	source entry/link
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410
NDUFS1	MITOCHONDRIAL COMPLEX I DEFICIENCY	OMIM	MIM:252010
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917

TUBB	CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS [...]	OMIM	MIM:615771	OMIM
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410	OMIM
NDUFS1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000	OMIM
NDUFS1	Mitochondrial Respiratory Chain Complex I Deficiency (nuclear genes)	DECIPHER	MIM:252010	OMIM
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886	orphanet
HLA-A	Birdshot chorioretinopathy	OrphaNet	OrphaNet:179	orphanet
NDUFS1	Isolated NADH-CoQ reductase deficiency	OrphaNet	OrphaNet:2609	orphanet
NDUFS1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241	orphanet
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	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

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PPP1R18	missense variant	ENST00000615527	?	ENSP00000480270	R/G	Agg/Ggg	?	?	1
PPP1R18	missense variant	ENST00000274853	NM_133471.3	ENSP00000274853	R/G	Agg/Ggg	?	?	1
PPP1R18	missense variant	ENST00000399199	NM_001134870.1	ENSP00000382150	R/G	Agg/Ggg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1654987	blood	3.60×10-6 (p-value)	MuTHER consortium	58
			adipocyte	2.42×10-6 (p-value)		
HLA-L	ENST00000463348	ILMN_1675282	blood	1.92×10-6 (p-value)	MuTHER consortium	47
ZNRD1	ENST00000471008	ILMN_2398587	blood	2.39×10-6 (p-value)	MuTHER consortium	44
ZNRD1	ENST00000359374					
ZNRD1-AS1_3	ENST00000611686					
ZNRD1	ENST00000332435					
ZNRD1	ENST00000376782					
ZNRD1	ENST00000376785					
ZNRD1	ENST00000463141					
TRIM31	ENST00000376734	ILMN_1748685	skin	4.22×10-6 (p-value)	MuTHER consortium	25
TRIM31	ENST00000484583					
TRIM31	ENST00000471695					
TRIM31	ENST00000468264					
HLA-V	ENST00000446817	ILMN_1660923	blood	1.32×10-9 (p-value)	MuTHER consortium	59
HLA-V	ENST00000429037		adipocyte	1.91×10-6 (p-value)	MuTHER consortium	59
			skin	4.10×10-6 (p-value)	MuTHER consortium	59
HCG22	ENST00000570223	ILMN_1667229	blood	2.57×10-6 (p-value)	MuTHER consortium	40
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					

GABBR1	ENST00000491829	ILMN_2395375	blood	1.94×10-5 (p-value)	MuTHER consortium	18
GABBR1	ENST00000377012					
GABBR1	ENST00000377034					
GABBR1	ENST00000478931					
GABBR1	ENST00000472823					
GABBR1	ENST00000377016					
VARS2	ENST00000321897	ILMN_1737585	skin	2.79×10-7 (p-value)	MuTHER consortium	36
VARS2	ENST00000625423					
VARS2	ENST00000541562					
VARS2	ENST00000542001					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
HCP5	ENST00000414046	ILMN_1803945	skin	5.42×10-9 (p-value)	MuTHER consortium	18
HCP5	ENST00000541196					
MICB	ENST00000538442	ILMN_1708006	skin	5.95×10-9 (p-value)	MuTHER consortium	18
MICB	ENST00000399150					
MICB	ENST00000252229					
IER3	ENST00000376377	ILMN_1682717	skin	2.84×10-5 (p-value)	MuTHER consortium	1
IER3	ENST00000259874					
ZFP57	?	ENSG00000204644	muscularis mucosae	6.78×10-8 (p-value)	GTEx Portal V6	82
PPP1R18	?	ENSG00000146112	muscularis mucosae	2.64×10-6 (p-value)	GTEx Portal V6	22
HLA-C	?	ENSG00000204525	muscularis mucosae	1.12×10-6 (p-value)	GTEx Portal V6	7
HLA-B	?	ENSG00000234745	muscularis mucosae	1.89×10-6 (p-value)	GTEx Portal V6	3
FLOT1	?	ENSG00000137312	lung	6.00×10-8 (p-value)	GTEx Portal V6	91
HLA-J	?	ENSG00000204622	lung	1.58×10-12 (p-value)	GTEx Portal V6	104
HLA-G	?	ENSG00000204632	lung	2.21×10-7 (p-value)	GTEx Portal V6	89
HLA-H	?	ENSG00000206341	lung	2.13×10-8 (p-value)	GTEx Portal V6	101
ZFP57	?	ENSG00000204644	lung	6.53×10-11 (p-value)	GTEx Portal V6	82
LINC00243	?	ENSG00000214894	lung	4.13×10-6 (p-value)	GTEx Portal V6	4
HLA-J	?	ENSG00000204622	atrial appendage	1.61×10-6 (p-value)	GTEx Portal V6	4
ZFP57	?	ENSG00000204644	atrial appendage	6.27×10-8 (p-value)	GTEx Portal V6	54
ZFP57	?	ENSG00000204644	transformed fibroblasts	3.56×10-8 (p-value)	GTEx Portal V6	81
HLA-J	?	ENSG00000204622	transformed fibroblasts	3.90×10-6 (p-value)	GTEx Portal V6	44
HLA-C	?	ENSG00000204525	transformed fibroblasts	7.51×10-8 (p-value)	GTEx Portal V6	75
HCG20	?	ENSG00000228022	transformed fibroblasts	2.40×10-7 (p-value)	GTEx Portal V6	10
HLA-H	?	ENSG00000206341	transformed fibroblasts	2.47×10-5 (p-value)	GTEx Portal V6	1
HLA-J	?	ENSG00000204622	blood	4.01×10-9 (p-value)	GTEx Portal V6	98
HLA-H	?	ENSG00000206341	blood	7.77×10-9 (p-value)	GTEx Portal V6	93
ZFP57	?	ENSG00000204644	blood	2.24×10-10 (p-value)	GTEx Portal V6	83
HLA-G	?	ENSG00000204632	blood	9.33×10-8 (p-value)	GTEx Portal V6	52
HLA-F-AS1	?	ENSG00000214922	blood	1.37×10-6 (p-value)	GTEx Portal V6	70

LINC00243	?	ENSG00000214894	blood	2.67×10-14 (p-value)	GTEx Portal V6	103
HLA-K	?	ENSG00000230795	blood	1.57×10-9 (p-value)	GTEx Portal V6	92
CCHCR1	?	ENSG00000204536	blood	4.75×10-6 (p-value)	GTEx Portal V6	6
HLA-C	?	ENSG00000204525	blood	8.58×10-7 (p-value)	GTEx Portal V6	16
VARS2	?	ENSG00000137411	blood	6.90×10-6 (p-value)	GTEx Portal V6	1
HLA-J	?	ENSG00000204622	breast	1.40×10-7 (p-value)	GTEx Portal V6	27
ZFP57	?	ENSG00000204644	breast	6.42×10-8 (p-value)	GTEx Portal V6	47
HLA-C	?	ENSG00000204525	breast	3.52×10-6 (p-value)	GTEx Portal V6	5
HLA-U	?	ENSG00000228078	tibial artery	5.57×10-7 (p-value)	GTEx Portal V6	72
HLA-J	?	ENSG00000204622	tibial artery	3.63×10-8 (p-value)	GTEx Portal V6	33
HLA-H	?	ENSG00000206341	tibial artery	5.15×10-7 (p-value)	GTEx Portal V6	39
ZFP57	?	ENSG00000204644	tibial artery	2.39×10-9 (p-value)	GTEx Portal V6	83
HLA-C	?	ENSG00000204525	tibial artery	1.92×10-9 (p-value)	GTEx Portal V6	82
HLA-J	?	ENSG00000204622	thyroid	6.77×10-7 (p-value)	GTEx Portal V6	39
ZFP57	?	ENSG00000204644	thyroid	2.65×10-10 (p-value)	GTEx Portal V6	83
ZNRD1	?	ENSG00000066379	thyroid	1.59×10-5 (p-value)	GTEx Portal V6	1
FLOT1	?	ENSG00000137312	thyroid	2.35×10-10 (p-value)	GTEx Portal V6	96
CCHCR1	?	ENSG00000204536	thyroid	8.47×10-10 (p-value)	GTEx Portal V6	93
PSORS1C1	?	ENSG00000204540	thyroid	5.65×10-7 (p-value)	GTEx Portal V6	11
PPP1R18	?	ENSG00000146112	thyroid	1.44×10-7 (p-value)	GTEx Portal V6	36
HLA-J	?	ENSG00000204622	skeletal muscle	1.24×10-9 (p-value)	GTEx Portal V6	103
HLA-H	?	ENSG00000206341	skeletal muscle	1.43×10-6 (p-value)	GTEx Portal V6	58
ZFP57	?	ENSG00000204644	skeletal muscle	1.78×10-6 (p-value)	GTEx Portal V6	23
HLA-C	?	ENSG00000204525	skeletal muscle	1.59×10-12 (p-value)	GTEx Portal V6	82
ZFP57	?	ENSG00000204644	liver	2.45×10-7 (p-value)	GTEx Portal V6	5
HLA-J	?	ENSG00000204622	sun exposed skin	2.03×10-7 (p-value)	GTEx Portal V6	20
HLA-H	?	ENSG00000206341	sun exposed skin	9.76×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	sun exposed skin	9.47×10-10 (p-value)	GTEx Portal V6	82
VARS2	?	ENSG00000137411	sun exposed skin	6.86×10-9 (p-value)	GTEx Portal V6	42
HLA-C	?	ENSG00000204525	sun exposed skin	3.64×10-7 (p-value)	GTEx Portal V6	26
ZFP57	?	ENSG00000204644	unexposed skin	1.63×10-6 (p-value)	GTEx Portal V6	4
ZFP57	?	ENSG00000204644	left ventricle	5.64×10-7 (p-value)	GTEx Portal V6	6
PSORS1C1	?	ENSG00000204540	left ventricle	3.69×10-7 (p-value)	GTEx Portal V6	9
HLA-H	?	ENSG00000206341	left ventricle	8.80×10-6 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	aorta	5.31×10-11 (p-value)	GTEx Portal V6	83
PPP1R18	?	ENSG00000146112	aorta	5.47×10-8 (p-value)	GTEx Portal V6	77
HLA-B	?	ENSG00000234745	aorta	6.92×10-6 (p-value)	GTEx Portal V6	1

HCG17	?	ENSG00000270604	subcutaneous adipocytes	3.78×10-8 (p-value)	GTEx Portal V6	70
ZFP57	?	ENSG00000204644	subcutaneous adipocytes	2.47×10-9 (p-value)	GTEx Portal V6	82
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	6.58×10-7 (p-value)	GTEx Portal V6	57
VARS2	?	ENSG00000137411	subcutaneous adipocytes	6.34×10-8 (p-value)	GTEx Portal V6	49
CCHCR1	?	ENSG00000204536	subcutaneous adipocytes	6.45×10-9 (p-value)	GTEx Portal V6	46
ZFP57	?	ENSG00000204644	stomach	7.56×10-7 (p-value)	GTEx Portal V6	65
HLA-C	?	ENSG00000204525	stomach	1.66×10-7 (p-value)	GTEx Portal V6	76
ZFP57	?	ENSG00000204644	visceral adipocytes	1.11×10-8 (p-value)	GTEx Portal V6	22
CCHCR1	?	ENSG00000204536	visceral adipocytes	2.13×10-6 (p-value)	GTEx Portal V6	7
HLA-H	?	ENSG00000206341	spleen	3.40×10-6 (p-value)	GTEx Portal V6	4
ZFP57	?	ENSG00000204644	tibial nerve	4.33×10-9 (p-value)	GTEx Portal V6	81
CCHCR1	?	ENSG00000204536	tibial nerve	1.48×10-7 (p-value)	GTEx Portal V6	77
PPP1R18	?	ENSG00000146112	tibial nerve	2.17×10-6 (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	tibial nerve	4.29×10-6 (p-value)	GTEx Portal V6	34
HLA-J	?	ENSG00000204622	esophagus mucosa	1.95×10-7 (p-value)	GTEx Portal V6	31
ZFP57	?	ENSG00000204644	esophagus mucosa	1.13×10-12 (p-value)	GTEx Portal V6	83
HLA-C	?	ENSG00000204525	esophagus mucosa	2.41×10-8 (p-value)	GTEx Portal V6	68
MICB	?	ENSG00000204516	esophagus mucosa	4.51×10-6 (p-value)	GTEx Portal V6	4
HLA-A	?	ENSG00000206503	brain	3.82×10-6 (q-value)	SeeQTL DB (Myers et al.)	4
?	?	ILMN_1679389	monocyte	1.31×10-14 (p-value)	Zeller et al.	8
?	?	ILMN_1666078	monocyte	1.81×10-37 (p-value)	Zeller et al.	8
?	?	ILMN_1671054	monocyte	1.64×10-45 (p-value)	Zeller et al.	8
HLA-G	ENST00000376828	ILMN_1656670	monocyte	3.91×10-19 (p-value)	Zeller et al.	8
HLA-G	ENST00000360323					
HLA-G	ENST00000478519					
HLA-G	ENST00000478355					
HLA-G	ENST00000428701					
HLA-G	ENST00000376815					
HLA-G	ENST00000376818					
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	1.68×10-40 (p-value)	Zeller et al.	8
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	2.53×10-24 (p-value)	Zeller et al.	8
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	8.29×10-28 (p-value)	Zeller et al.	8
BTN3A2	ENST00000528541					
BTN3A3	ENST00000467524					
BTN3A3	ENST00000482451					
BTN3A3	ENST00000494393					
BTN3A2	ENST00000527417					
BTN3A3	ENST00000244519					
BTN3A2	ENST00000528222					
BTN3A3	ENST00000474790					
BTN3A2	ENST00000377708					

BTN3A3	ENST00000496719					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000527422					
BTN3A2	ENST00000356386					
?	?	ILMN_1883997	monocyte	1.74×10-44 (p-value)	Zeller et al.	8
HLA-C	ENST00000470363	ILMN_2150787	monocyte	3.78×10-23 (p-value)	Zeller et al.	7
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
HLA-C	ENST00000466892					
?	?	ILMN_1893395	monocyte	1.16×10-14 (p-value)	Zeller et al.	1
HIST1H3I	ENST00000616365	ILMN_2207865	b-cell	1.43×10-4 (p-value)	Fairfax et al.	8
ZNRD1	ENST00000471008	ILMN_2398587	b-cell	2.62×10-4 (p-value)	Fairfax et al.	2
ZNRD1	ENST00000359374					
ZNRD1-AS1_3	ENST00000611686					
ZNRD1	ENST00000376785					
ZNRD1	ENST00000376782					
ZNRD1	ENST00000332435					
ZNRD1	ENST00000463141					
C2	ENST00000486124	ILMN_1710740	monocyte	7.85×10-6 (p-value)	Fairfax et al.	2
C2	ENST00000469372					
CFB	ENST00000425368					
C2	ENST00000497706					
C2	ENST00000442278					
C2	ENST00000452323					
C2	ENST00000383177					
C2	ENST00000299367					
C2	ENST00000482060					
?	?	ILMN_1679389	b-cell	4.36×10-6 (p-value)	Fairfax et al.	17
IER3	ENST00000376377	ILMN_1682717	monocyte	2.83×10-5 (p-value)	Fairfax et al.	20
IER3	ENST00000259874					
RPP21	ENST00000489124	ILMN_2186597	b-cell	4.61×10-4 (p-value)	Fairfax et al.	1
RPP21	ENST00000466327					
RPP21	ENST00000436442					
TRIM39-RPP21	ENST00000623385					
RPP21	ENST00000473266					
TRIM39-RPP21	ENST00000513556					
RPP21	ENST00000498414					
RPP21	ENST00000433076					
RPP21	ENST00000442966					
RPP21	ENST00000491477					
RPP21	ENST00000428040					
OR5V1	ENST00000543825	ILMN_1683352	b-cell	3.80×10-6 (p-value)	Fairfax et al.	10
OR5V1	ENST00000377154					
VARS2	ENST00000321897	ILMN_1737585	monocyte	5.36×10-7 (p-value)	Fairfax et al.	2
VARS2	ENST00000625423		b-cell	5.12×10-5 (p-value)	Fairfax et al.	2
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					

VARS2	ENST00000477288						
VARS2	ENST00000473916						
VARS2	ENST00000476162						
ZFP57	?	ENSG00000204644	transverse colon	2.63×10-6 (p-value)	GTEx Portal V6	2
HLA-C	?	ENSG00000204525	transverse colon	2.66×10-8 (p-value)	GTEx Portal V6	66
TRIM26BP	?	ENSG00000236475	transverse colon	2.59×10-6 (p-value)	GTEx Portal V6	19
CCHCR1	?	ENSG00000204536	transverse colon	1.82×10-6 (p-value)	GTEx Portal V6	3
ZFP57	?	ENSG00000204644	coronary artery	2.82×10-6 (p-value)	GTEx Portal V6	2
HLA-J	?	ENSG00000204622	coronary artery	2.14×10-6 (p-value)	GTEx Portal V6	20
HLA-C	ENST00000470363	216526_x_at	blood	1.50×10-8 (p-value)	Dixon et al.	4
HLA-C	ENST00000383329					
HLA-C	ENST00000487245					
HLA-C	ENST00000376228					
HLA-C	ENST00000466892					
HLA-C	ENST00000376237					
HLA-C	ENST00000620806					
RNF39	?	ENSG00000204618	pancreas	2.26×10-6 (p-value)	GTEx Portal V6	1
WASF5P	?	ENSG00000231402	pancreas	3.25×10-8 (p-value)	GTEx Portal V6	42
HLA-C	?	ENSG00000204525	pancreas	1.65×10-6 (p-value)	GTEx Portal V6	4
MICB	?	ENSG00000204516	cerebellar hemisphere	1.84×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	cerebellum	1.25×10-6 (p-value)	GTEx Portal V6	1
HLA-G	?	ENSG00000204632	EBV lymphocytes	1.79×10-6 (p-value)	GTEx Portal V6	4
HCG17	?	ENSG00000270604	hypothalamus	1.06×10-6 (p-value)	GTEx Portal V6	4

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
HLA-A	?	ENSG00000206503	chr6	brain	6.61×10-4 (q-value)	SeeQTL DB (Myers et al.)	4
?	?	ILMN_1752592	chr6	monocyte	8.64×10-29 (p-value)	Zeller et al.	8
HLA-C	ENST00000470363	ILMN_2150787	chr6	monocyte	2.91×10-19 (p-value)	Zeller et al.	1
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						
?	?	ILMN_1893395	chr6	monocyte	1.06×10-12 (p-value)	Zeller et al.	4
?	?	ILMN_1715169	chr6	monocyte	9.75×10-15 (p-value)	Zeller et al.	6
?	?	ILMN_1661266	chr6	monocyte	4.95×10-15 (p-value)	Zeller et al.	6
TMEM154	ENST00000304385	ILMN_2088124	chr4	blood	1.16×10-10 (p-value)	Westra et al.	1
NDUFS1	ENST00000455934	ILMN_1728810	chr2	blood	5.94×10-8 (p-value)	Westra et al.	1
NDUFS1	ENST00000233190						
NDUFS1	ENST00000423725						
BTN3A2	ENST00000524682	209846_s_at	chr6	blood	3.10×10-11 (p-value)	Dixon et al.	6
BTN3A2	ENST00000604202						

BTN3A2	ENST00000396934						
BTN3A2	ENST00000532294						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000377708						
BTN3A2	ENST00000527422						
BTN3A2	ENST00000531055						
BTN3A2	ENST00000396948						
BTN3A2	ENST00000356386						
BTN3A2	ENST00000396934	212613_at	chr6	blood	1.40×10-8 (p-value)	Dixon et al.	3
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000244519	204820_s_at	chr6	blood	3.20×10-10 (p-value)	Dixon et al.	5
BTN3A2	ENST00000396934						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000480110						
HLA-C	ENST00000470363	216526_x_at	chr6	blood	3.20×10-8 (p-value)	Dixon et al.	1
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000466892						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
JUND	MA0491.1	8	yes	0.000	1
FOSL1	MA0477.1	4	yes	0.000	1
SP1	MA0079.3	10	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000047900		HCG19P
		HCG19P
		HCG19P
		HCG19P
		HCG19P
		HCG19P
ENCP00000047941		FLOT1
		FLOT1
		FLOT1
		FLOT1
		FLOT1
		FLOT1
		FLOT1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444288	1	ENCP00000047932	NRM
			NRM

			NRM NRM NRM NRM NRM
		ENCP00000047889	TRIM26 TRIM26 TRIM26 TRIM26 TRIM26 TRIM26 TRIM26 TRIM26
ENCE00000444247	1	ENCP00000047877	RNF39 RNF39 RNF39 RNF39 RNF39 RNF39 RNF39 RNF39
		ENCP00000047957	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
		ENCP00000047879	TRIM31 TRIM31 TRIM31 TRIM31 TRIM31 TRIM31 TRIM31 TRIM31
ENCE00000444553	1	ENCP00000047941	FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1 FLOT1
ENCE00000444465	1	ENCP00000047921	ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1 ATAT1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487584 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00000487598 (promoter flanking region)	2	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	FOSL2, Jund, USF1, H3K4me1, H3K4me2, HNF4A, FOXA1
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1, H3K27me3

		nervous (NH-A)	DNase1, H3K27me3
ENSR00001702960 (enhancer)	1	liver (HepG2)	H3K36me3
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001702961 (enhancer)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF
		HSMMtube	H3K27me3, CTCF
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	CTCF, H3K27me3
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, H3K36me3
		blood (GM12878)	CTCF
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF, H3K27me3
		NHLF	CTCF, H3K27me3
		Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		A549	CTCF
ENSR00000487633 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001494491 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K4me2, Rad21, Nrsf, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF, H3K27me3, DNase1
		blood (GM12878)	DNase1, Rad21, CTCF
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	H3K27me3, CTCF
		skin (NHEK)	CTCF, H3K27me3, DNase1
		NHLF	CTCF, H3K27me3
		Osteobl	CTCF, H3K27me3
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	Nrsf, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, DNase1, H3K27me3, CTCF
		A549	H3K27me3, CTCF
ENSR00001494494 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	DNase1, H3K27me3
		breast (HMEC)	DNase1
ENSR00001032027 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	DNase1, H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	DNase1
		liver (HepG2)	H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1, H3K27me3
		skin (NHEK)	H3K27me3, DNase1
		NHLF	DNase1, H3K27me3

		Osteobl	H3K27me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27me3
		cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me1, H3K4me3, H3K27me3
		endothelium (HUVEC)	H3K27me3
		A549	H3K27me3
ENSR00000487656 e! (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		HSMMtube	H3K27me3, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		liver (HepG2)	DNase1, H3K27ac, Cmyc, FOXA1, HNF4A, Jund, HNF4G, HDAC2, H3K4me1, p300
		lung (IMR90)	DNase1
		blood (GM12878)	DNase1, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K27ac
		NHLF	H3K27ac, DNase1
		Osteobl	H3K4me2, H3K27ac
		breast (HMEC)	DNase1, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K27me3
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Max, DNase1, H3K27ac, Cjun
		A549	DNase1
ENSR00000487671 e! (promoter)	2	embryonic stem cell (H1ESC)	DNase1, TAF1, H3K4me2, SP1, Srf, H3K4me3, HDAC2, CTCF, H3K27ac, H3K36me3, H3K27me3, PolII, Rad21, Egr1, Jund, Sin3Ak20, H3K9ac
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, CTCF, H3K79me2
		blood (K562)	Ini1, Egr1, CTCF, H3K36me3, DNase1, H3K4me3, H3K4me2, PolII, FOSL1, H3K27ac, Max, ZBTB7A, Srf, H3K4me1, H3K9ac, Gabp, Cmyc, H3K79me2, HEY1, H2AZ, Rad21, TAF1, ELF1
		skin (NHDF-AD)	H3K36me3, H3K4me1, CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1, H2AZ
		liver (HepG2)	DNase1, Gabp, PolII, Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3
		lung (IMR90)	DNase1, H4K8ac, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K56ac, CTCF
		blood (GM12878)	ELF1, Rad21, H3K79me2, H3K4me3, Pax5, H3K27ac, H3K4me2, CTCF, H3K9ac, H3K36me3, Gabp, Yy1, EBF1, PolII, H2AZ, Tcf12, DNase1, Egr1
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K36me3, DNase1, CTCF, H3K4me3, H3K4me2, H3K4me1, H3K27ac, H3K9ac
		NHLF	DNase1, H3K4me3, H3K4me1, CTCF, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K9ac, H3K4me3, H3K36me3, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	DNase1, Jund, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, CTCF, H3K36me3, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K27ac, PolII, CTCF, DNase1, Cmyc, H3K9ac, H3K4me3, H3K4me2, Cjun, Max
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000487679 e! (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K27me3, PolII, Rad21, Egr1, Jund, TAF7, Sin3Ak20, CTCF, SIX5, H3K27ac, ATF3, Yy1, TAF1, H3K4me2, SP1, USF1, H3K36me3, H3K9ac, H3K4me3
		HSMMtube	H3K36me3, H2AZ, H3K4me3, CTCF, H3K27ac, H3K4me2, H3K9ac, DNase1
		blood (K562)	H3K27ac, H3K36me3, DNase1, H3K4me3, CTCF, H3K4me2, Max, SP2, PU1, H3K9ac, USF1, H3K79me2, HEY1, H2AZ, Rad21, TAF1, Cfos, CTCFL, Cmyc, PolII, H4K20me1, H3K27me3, Yy1, Ini1, Brg1, Egr1
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K36me3, H3K4me1, CTCF, H4K20me1, H3K4me3, DNase1, H3K9ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, Rad21, TAF1, USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3, H3K79me2, PolII, ATF3, Yy1, H4K20me1
		lung (IMR90)	DNase1, H4K8ac, H4K20me1, H3K79me2, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K56ac, CTCF
		blood (GM12878)	Pbx3, TAF1, ATF3, SIX5, PolII, BCLAF1, H2AZ, Tcf12, DNase1, Egr1, Yy1, ELF1, Rad21, H3K79me2, H3K4me3, USF1, H3K27ac, Cfos, H3K4me2, CTCF, H3K9ac, H3K36me3
		nervous (NH-A)	DNase1, H3K9ac, H4K20me1, CTCF, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1
		NHLF	H3K36me3, H3K27ac, H3K9ac, DNase1, H3K4me3, H3K4me1, CTCF
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2, CTCF, H3K36me3
		blood (DND-41)	H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	H3K36me3, CTCF, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K9ac, Nrsf, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, PolII, Cmyc, CTCF, Ini1, H3K36me3

		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, CTCF, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000487687 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		HSMMtube	H3K36me3
		blood (K562)	H3K27me3, PU1, H3K36me3
		skin (NHDF-AD)	H3K36me3
		muscle (HSMM)	H3K36me3
		liver (HepG2)	DNase1, H3K4me1, H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	Yy1, PU1, PolII, H3K4me1, DNase1, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K4me1, H3K36me3
		cervix (HeLa-S3)	H3K27ac, PolII
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me1
		endothelium (HUVEC)	PolII, H3K36me3, DNase1
		A549	H3K36me3
ENSR00000487711 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		liver (HepG2)	H3K4me1, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me2
		blood (GM12878)	H2AZ, DNase1, H3K27ac
		lung (IMR90)	DNase1, H3K27ac
		blood (DND-41)	H3K4me1, H3K27ac
		blood (K562)	DNase1, H2AZ
ENSR00000487714 (promoter flanking region)	1	endothelium (HUVEC)	DNase1
		liver (HepG2)	H3K4me1, FOXA1
		blood (K562)	H3K4me1
ENSR00000662312 (promoter flanking region)	2	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, PolII, TAF7
		Osteobl	H3K27ac
		blood (K562)	USF1, HEY1, H2AZ, Cjun, Cfos, PolII, MEF2A, Max, Ini1, Brg1, DNase1, FOSL1, H3K27ac
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, DNase1
		muscle (HSMM)	H2AZ
		cervix (HeLa-S3)	DNase1, PolII, TAF1, H3K9ac, Jund, H3K27ac, H3K4me1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me1, H3K27ac, PolII, DNase1, Cmyc
		liver (HepG2)	DNase1, FOSL2, Jund, PolII, H3K4me1
		lung (IMR90)	DNase1, H2BK12ac, H3K18ac, H2BK120ac, H2BK20ac, H4K91ac
		blood (GM12878)	EBF1, PolII, BATF, DNase1, IRF4, BCL3, BCL11A, Pax5, NFKB
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, H3K27ac, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ZNRD1	1	ENST00000332435 ENST00000359374 ENST00000376782 ENST00000376785 ENST00000463141 ENST00000471008	hsa-miR-199a-5p hsa-miR-199b-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABCF1	upstream gene variant	2298	ENST00000376545	NM_001090.2	ENSP00000365728	1
ABCF1	upstream gene variant	2338	ENST00000441867	?	ENSP00000405512	1
ABCF1	upstream gene variant	1503	ENST00000542772	?	?	1
ABCF1	upstream gene variant	2281	ENST00000326195	NM_001025091.1	ENSP00000313603	1

ABCF1	upstream gene variant	2347	ENST00000468958	?	ENSP00000440893	1
ABCF1	downstream gene variant	1451	ENST00000475993	?	ENSP00000445100	1
AL662797.1	upstream gene variant, downstream gene variant	112	ENST00000620079	?	?	3
AL662800.2	downstream gene variant	2370	ENST00000583820	?	?	1
ATAT1	downstream gene variant	4727	ENST00000462304	?	?	1
ATAT1	downstream gene variant	4306	ENST00000330083	NM_001031722.2	ENSP00000327832	1
ATAT1	downstream gene variant	4306	ENST00000376485	?	ENSP00000365668	1
ATAT1	downstream gene variant	4306	ENST00000471782	?	?	1
ATAT1	downstream gene variant	4306	ENST00000479562	?	?	1
ATAT1	downstream gene variant	2875	ENST00000493388	?	?	1
ATAT1	downstream gene variant	4306	ENST00000468713	?	?	1
C6orf136	upstream gene variant	137	ENST00000467801	?	ENSP00000419610	1
C6orf136	upstream gene variant	3482	ENST00000488383	?	ENSP00000419968	1
C6orf136	upstream gene variant	3848	ENST00000446773	?	ENSP00000415810	1
C6orf136	upstream gene variant	3466	ENST00000484551	?	ENSP00000418599	1
C6orf136	upstream gene variant	3553	ENST00000463794	?	ENSP00000418223	1
C6orf136	upstream gene variant	255	ENST00000468785	?	ENSP00000419460	1
C6orf136	upstream gene variant	3591	ENST00000376471	NM_145029.3	ENSP00000365654	1
C6orf136	upstream gene variant	3466	ENST00000293604	NM_001161376.1	ENSP00000293604	1
C6orf136	upstream gene variant	3500	ENST00000376473	NM_001109938.2	ENSP00000365656	1
C6orf136	upstream gene variant	1482	ENST00000460172	?	?	1
C6orf136	upstream gene variant, downstream gene variant	1216	ENST00000493705	?	?	2
DHX16	upstream gene variant	3372	ENST00000415603	?	ENSP00000399101	1
DHX16	downstream gene variant	1990	ENST00000376437	?	ENSP00000365620	1
DHX16	downstream gene variant, upstream gene variant	1990	ENST00000376442	NM_001164239.1, NM_003587.4	ENSP00000365625	2
FLOT1	upstream gene variant	384	ENST00000476729	?	?	2
FLOT1	downstream gene variant	1925	ENST00000454845	?	ENSP00000391341	1
FLOT1	downstream gene variant	176	ENST00000484693	?	?	2
FLOT1	downstream gene variant	1577	ENST00000418160	?	ENSP00000404300	1
FLOT1	downstream gene variant	1670	ENST00000484168	?	?	1
FLOT1	upstream gene variant	325	ENST00000487376	?	?	2
FLOT1	downstream gene variant	1592	ENST00000445853	?	ENSP00000398834	1
FLOT1	downstream gene variant	1671	ENST00000416018	?	ENSP00000412058	1
FLOT1	downstream gene variant	1574	ENST00000470643	?	?	1
GNL1	downstream gene variant	4911	ENST00000487166	?	?	1
GNL1	downstream gene variant	198	ENST00000376621	NM_005275.3	ENSP00000365806	2

HCG17	upstream gene variant	1993	ENST00000455957	?	?	2
HCG17	upstream gene variant	2342	ENST00000453558	?	?	1
HCG18	downstream gene variant	3783	ENST00000602591	?	?	1
HCG18	upstream gene variant	1610	ENST00000412685	?	?	2
HCG18	upstream gene variant	2100	ENST00000454129	?	?	1
HCG18	upstream gene variant	2089	ENST00000438412	?	?	1
HCG18	upstream gene variant	4008	ENST00000602516	?	?	2
HCG18	upstream gene variant	2113	ENST00000602290	?	?	1
HCG18	upstream gene variant	1579	ENST00000413358	?	?	2
HCG18	upstream gene variant	2129	ENST00000449544	?	?	1
HCG18	upstream gene variant	2072	ENST00000602550	?	?	1
HCG18	upstream gene variant	1326	ENST00000426882	?	?	2
HCG18	upstream gene variant	2339	ENST00000602498	?	?	1
HCG18	upstream gene variant	2142	ENST00000444126	?	?	1
HCG18	upstream gene variant	2090	ENST00000454269	?	?	1
HCG19P	downstream gene variant, upstream gene variant	151	ENST00000448756	?	?	14
HCG20	downstream gene variant	1545	ENST00000439406	?	?	1
HCG20	upstream gene variant, downstream gene variant	1545	ENST00000422944	?	?	2
HCG4P3	downstream gene variant	2938	ENST00000458060	?	?	1
HLA-J	upstream gene variant	3413	ENST00000495278	?	?	1
HLA-J	upstream gene variant	3404	ENST00000462773	?	?	1
HLA-J	upstream gene variant	3423	ENST00000494367	?	?	1
HLA-J	upstream gene variant	3400	ENST00000469281	?	?	1
HLA-L	upstream gene variant	2391	ENST00000482052	?	?	1
HLA-L	upstream gene variant, downstream gene variant	1288	ENST00000420110	?	?	2
HLA-L	upstream gene variant	2404	ENST00000463348	?	?	1
HLA-N	upstream gene variant, downstream gene variant	39	ENST00000437516	?	?	3
IER3	downstream gene variant, upstream gene variant	1619	ENST00000376377	?	ENSP00000365557	4
IER3	downstream gene variant, upstream gene variant	1619	ENST00000259874	NM_003897.3	ENSP00000259874	4
LINC00243	downstream gene variant	4859	ENST00000419357	?	?	1
LINC00243	downstream gene variant	75	ENST00000399196	?	?	1
MDC1	upstream gene variant	3623	ENST00000416571	?	ENSP00000400979	1
MDC1	upstream gene variant	3351	ENST00000425072	?	ENSP00000396989	1
MDC1	upstream gene variant	2909	ENST00000376406	NM_014641.2	ENSP00000365588	1
MDC1	upstream gene variant	3723	ENST00000613547	?	ENSP00000483601	1
MDC1	upstream gene variant	3677	ENST00000422266	?	ENSP00000411310	1
MICC	upstream gene variant	896	ENST00000445710	?	?	2

MRPS18B	downstream gene variant	2249	ENST00000472229	?	?	1
MRPS18B	downstream gene variant	2249	ENST00000472267	?	?	1
MRPS18B	downstream gene variant	2249	ENST00000259873	NM_014046.3	ENSP00000259873	1
NRM	downstream gene variant	3043	ENST00000444096	?	ENSP00000397892	1
NRM	downstream gene variant	3043	ENST00000259953	NM_007243.2	ENSP00000259953	1
NRM	downstream gene variant	3043	ENST00000376420	NM_001270709.1	ENSP00000365602	1
NRM	downstream gene variant	3043	ENST00000470733	?	?	1
NRM	downstream gene variant	3043	ENST00000462857	?	?	1
NRM	downstream gene variant	3046	ENST00000376421	NM_001270707.1	ENSP00000365603	1
NRM	downstream gene variant	3043	ENST00000495946	?	?	1
NRM	downstream gene variant	3043	ENST00000482141	?	?	1
NRM	downstream gene variant	3043	ENST00000474864	?	?	1
PAIP1P1	downstream gene variant	1212	ENST00000446875	?	?	1
PPP1R10	downstream gene variant	1770	ENST00000376511	NM_002714.3	ENSP00000365694	1
PPP1R10	downstream gene variant	4943	ENST00000496955	?	?	1
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376772	NM_021959.2	ENSP00000365963	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376758	?	ENSP00000365949	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376763	?	ENSP00000365954	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376765	?	ENSP00000365956	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376769	?	ENSP00000365960	2
PPP1R11	upstream gene variant, downstream gene variant	537	ENST00000376773	?	ENSP00000365964	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000399199	NM_001134870.1	ENSP00000382150	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000488324	?	?	2
PPP1R18	downstream gene variant, upstream gene variant	31	ENST00000615527	?	ENSP00000480270	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000274853	NM_133471.3	ENSP00000274853	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000467662	?	?	2
PPP1R18	downstream gene variant, upstream gene variant	29	ENST00000615892	?	ENSP00000482578	3
PTMAP1	upstream gene variant, downstream gene variant	1844	ENST00000455552	?	?	2
RN7SL353P	upstream gene variant, downstream gene variant	1557	ENST00000579902	?	?	5
RPP21	downstream gene variant	3693	ENST00000489124	?	?	2
RPP21	downstream gene variant	3693	ENST00000436442	NM_001199121.1	ENSP00000397778	2
RPP21	downstream gene variant	3693	ENST00000428040	?	ENSP00000394320	2
RPP21	downstream gene variant	3693	ENST00000498414	?	?	2
RPP21	downstream gene variant	3693	ENST00000433076	NM_001199120.1	ENSP00000409799	2
RPP21	downstream gene variant	3666	ENST00000491477	?	?	2
RPP21	downstream gene variant	4057	ENST00000466327	?	?	2
RPP21	downstream gene variant	3693	ENST00000442966	NM_024839.2	ENSP00000403833	2

RPP21	downstream gene variant	3693	ENST00000473266	?	?	2
SUCLA2P1	downstream gene variant	3275	ENST00000425839	?	?	1
TRIM10	upstream gene variant	238	ENST00000376704	NM_052828.2	ENSP00000365894	1
TRIM10	upstream gene variant	238	ENST00000449742	NM_006778.3	ENSP00000397073	1
TRIM15	upstream gene variant	2034	ENST00000619857	?	ENSP00000484001	2
TRIM15	upstream gene variant	703	ENST00000477944	?	?	1
TRIM15	upstream gene variant	2044	ENST00000376694	NM_033229.2	ENSP00000365884	2
TRIM15	upstream gene variant	2513	ENST00000376688	?	ENSP00000365878	2
TRIM26	upstream gene variant	996	ENST00000437089	?	ENSP00000395491	1
TRIM26	downstream gene variant	4732	ENST00000418026	?	ENSP00000387530	1
TRIM26	downstream gene variant	2777	ENST00000416596	?	ENSP00000413673	1
TRIM26	downstream gene variant	4979	ENST00000434785	?	ENSP00000400920	1
TRIM26	upstream gene variant	3365	ENST00000480999	?	?	1
TRIM26BP	upstream gene variant	64	ENST00000427723	?	?	2
TRIM31	upstream gene variant	3745	ENST00000485864	?	?	1
TRIM31	upstream gene variant	2825	ENST00000376734	NM_007028.3	ENSP00000365924	1
TRIM31	downstream gene variant, upstream gene variant	3382	ENST00000493404	?	?	2
TRIM31	downstream gene variant, upstream gene variant	910	ENST00000480808	?	?	2
TRIM31	upstream gene variant	2396	ENST00000484583	?	?	1
TRIM31	upstream gene variant	2179	ENST00000471695	?	?	1
TRIM31-AS1	downstream gene variant	1207	ENST00000440874	?	?	1
TRIM39	upstream gene variant	1053	ENST00000420746	?	ENSP00000388224	1
TRIM39	downstream gene variant	1717	ENST00000458516	?	ENSP00000405928	1
TRIM39	downstream gene variant	1973	ENST00000428404	?	ENSP00000405498	1
TRIM39	downstream gene variant	1929	ENST00000440271	?	ENSP00000394768	1
TRIM39	upstream gene variant	413	ENST00000396547	?	ENSP00000379796	1
TRIM39	downstream gene variant	2006	ENST00000428555	?	ENSP00000397952	1
TRIM39-RPP21	upstream gene variant, downstream gene variant	431	ENST00000623385	?	ENSP00000485378	3
TRIM39-RPP21	upstream gene variant, downstream gene variant	835	ENST00000513556	NM_001199119.1	ENSP00000424048	3
TUBB	upstream gene variant	869	ENST00000396389	?	ENSP00000379672	1
TUBB	upstream gene variant	776	ENST00000330914	NM_001293215.1	ENSP00000365578	1
TUBB	upstream gene variant	901	ENST00000396384	?	ENSP00000379668	1
UBQLN1P1	downstream gene variant, upstream gene variant	152	ENST00000441056	?	?	16
XXbac-BPG252P9.10	upstream gene variant	1349	ENST00000607333	?	?	3
XXbac-BPG252P9.9	downstream gene variant	2307	ENST00000607476	?	?	1
XXbac-BPG283O16.9	downstream gene variant	1415	ENST00000624252	?	?	1
Y_RNA	downstream gene variant, upstream gene variant	2170	ENST00000365118	?	?	3

ZNRD1-AS1	upstream gene variant	3566	ENST00000431012	?	?	1
ZNRD1-AS1	upstream gene variant	3601	ENST00000376797	?	?	1
ZNRD1-AS1	upstream gene variant	3817	ENST00000425604	?	?	1
ZNRD1-AS1	upstream gene variant	3556	ENST00000421692	?	?	1
ZNRD1-AS1	upstream gene variant	3766	ENST00000452229	?	?	1
ZNRD1-AS1	downstream gene variant, upstream gene variant	1223	ENST00000437417	?	?	2
ZNRD1-AS1	upstream gene variant	3602	ENST00000448093	?	?	1
ZNRD1-AS1	upstream gene variant	3561	ENST00000420251	?	?	1
ZNRD1-AS1_3	downstream gene variant	3562	ENST00000611686	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
GNL1	ENST00000376621	NM_005275.3	ENSP00000365806	A	gcG/gcA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABCF1	ENST00000326195	NM_001025091.1	ENSP00000313603	1
ABCF1	ENST00000468958	?	ENSP00000440893	1
ABCF1	ENST00000441867	?	ENSP00000405512	1
ABCF1	ENST00000376545	NM_001090.2	ENSP00000365728	1
ATAT1	ENST00000376485	?	ENSP00000365668	3
ATAT1	ENST00000493388	?	?	2
ATAT1	ENST00000318999	?	ENSP00000324222	3
ATAT1	ENST00000468713	?	?	3
ATAT1	ENST00000376483	?	ENSP00000365666	3
ATAT1	ENST00000330083	NM_001031722.2	ENSP00000327832	3
ATAT1	ENST00000319027	NM_001254952.1	ENSP00000324459	3
ATAT1	ENST00000471782	?	?	3
ATAT1	ENST00000329992	NM_024909.2	ENSP00000332374	3
ATAT1	ENST00000479562	?	?	1
ATAT1	ENST00000462304	?	?	3
C6orf136	ENST00000376473	NM_001109938.2	ENSP00000365656	1
C6orf136	ENST00000465699	?	ENSP00000418154	1
C6orf136	ENST00000293604	NM_001161376.1	ENSP00000293604	1
C6orf136	ENST00000488383	?	ENSP00000419968	1
C6orf136	ENST00000376471	NM_145029.3	ENSP00000365654	1
C6orf136	ENST00000446773	?	ENSP00000415810	1
C6orf136	ENST00000487873	?	ENSP00000418616	1
C6orf136	ENST00000463794	?	ENSP00000418223	1

C6orf136	ENST00000484551	?	ENSP00000418599	1
FLOT1	ENST00000418160	?	ENSP00000404300	2
FLOT1	ENST00000416018	?	ENSP00000412058	2
FLOT1	ENST00000470643	?	?	2
FLOT1	ENST00000484168	?	?	2
FLOT1	ENST00000476729	?	?	2
FLOT1	ENST00000413165	?	ENSP00000395333	4
FLOT1	ENST00000376389	NM_005803.2	ENSP00000365569	4
FLOT1	ENST00000454845	?	ENSP00000391341	2
FLOT1	ENST00000438162	?	ENSP00000400615	4
FLOT1	ENST00000445853	?	ENSP00000398834	2
FLOT1	ENST00000487376	?	?	2
HCG17	ENST00000453558	?	?	11
HCG18	ENST00000454129	?	?	4
HCG18	ENST00000444126	?	?	4
HCG18	ENST00000602550	?	?	4
HCG18	ENST00000438412	?	?	4
HCG18	ENST00000412685	?	?	4
HCG18	ENST00000454269	?	?	4
HCG18	ENST00000426882	?	?	4
HCG18	ENST00000449544	?	?	4
HCG18	ENST00000413358	?	?	4
HCG18	ENST00000602498	?	?	4
HCG18	ENST00000602290	?	?	4
HCG20	ENST00000439406	?	?	2
HLA-L	ENST00000463348	?	?	1
HLA-L	ENST00000482052	?	?	3
LINC00243	ENST00000419357	?	?	4
LINC00243	ENST00000399196	?	?	2
MICC	ENST00000445710	?	?	1
NRM	ENST00000474864	?	?	1
NRM	ENST00000470733	?	?	1
NRM	ENST00000444096	?	ENSP00000397892	1
NRM	ENST00000376421	NM_001270707.1	ENSP00000365603	1
NRM	ENST00000482141	?	?	1
NRM	ENST00000495946	?	?	1
NRM	ENST00000259953	NM_007243.2	ENSP00000259953	1
NRM	ENST00000462857	?	?	1
NRM	ENST00000376420	NM_001270709.1	ENSP00000365602	1
PPP1R18	ENST00000274853	NM_133471.3	ENSP00000274853	1
PPP1R18	ENST00000467662	?	?	2
PPP1R18	ENST00000399199	NM_001134870.1	ENSP00000382150	1

PPP1R18	ENST00000615892	?	ENSP00000482578	1
PPP1R18	ENST00000615527	?	ENSP00000480270	1
PPP1R18	ENST00000488324	?	?	2
TRIM10	ENST00000449742	NM_006778.3	ENSP00000397073	2
TRIM10	ENST00000376704	NM_052828.2	ENSP00000365894	2
TRIM15	ENST00000376694	NM_033229.2	ENSP00000365884	1
TRIM15	ENST00000619857	?	ENSP00000484001	1
TRIM15	ENST00000433744	?	ENSP00000398285	1
TRIM15	ENST00000376688	?	ENSP00000365878	1
TRIM26	ENST00000418026	?	ENSP00000387530	2
TRIM26	ENST00000434785	?	ENSP00000400920	2
TRIM26	ENST00000487829	?	?	2
TRIM26	ENST00000480999	?	?	1
TRIM26	ENST00000453195	NM_001242783.1	ENSP00000391879	4
TRIM26	ENST00000437089	?	ENSP00000395491	3
TRIM26	ENST00000454678	NM_003449.4	ENSP00000410446	4
TRIM26	ENST00000416596	?	ENSP00000413673	2
TRIM31	ENST00000468264	?	?	1
TRIM31	ENST00000485864	?	?	1
TRIM31	ENST00000376734	NM_007028.3	ENSP00000365924	1
TRIM31-AS1	ENST00000440874	?	?	1
TRIM39	ENST00000458516	?	ENSP00000405928	1
TRIM39	ENST00000396547	?	ENSP00000379796	1
TRIM39	ENST00000396548	?	ENSP00000379797	2
TRIM39	ENST00000428728	?	ENSP00000406019	2
TRIM39	ENST00000428404	?	ENSP00000405498	1
TRIM39	ENST00000428555	?	ENSP00000397952	1
TRIM39	ENST00000396551	?	ENSP00000379800	2
TRIM39	ENST00000376659	NM_172016.2	ENSP00000365847	2
TRIM39	ENST00000420746	?	ENSP00000388224	1
TRIM39	ENST00000440271	?	ENSP00000394768	1
TRIM39	ENST00000376656	NM_021253.3	ENSP00000365844	2
TRIM39-RPP21	ENST00000513556	NM_001199119.1	ENSP00000424048	1
TRIM39-RPP21	ENST00000623385	?	ENSP00000485378	1
TRIM40	ENST00000376724	?	ENSP00000365914	1
TRIM40	ENST00000307859	NM_138700.4	ENSP00000308310	1
TRIM40	ENST00000396581	NM_001286633.1	ENSP00000379826	1
TUBB	ENST00000327892	NM_178014.2	ENSP00000339001	1
ZNRD1-AS1	ENST00000425604	?	?	2
ZNRD1-AS1	ENST00000420251	?	?	2
ZNRD1-AS1	ENST00000376797	?	?	2
ZNRD1-AS1	ENST00000448093	?	?	2

ZNRD1-AS1	ENST00000448093	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATAT1	ENST00000318999	?	ENSP00000324222	1
ATAT1	ENST00000376483	?	ENSP00000365666	1
ATAT1	ENST00000319027	NM_001254952.1	ENSP00000324459	1
ATAT1	ENST00000329992	NM_024909.2	ENSP00000332374	1
RNF39	ENST00000376751	NM_170769.2	ENSP00000365942	1
RNF39	ENST00000244360	NM_025236.3	ENSP00000244360	1
TRIM26	ENST00000453195	NM_001242783.1	ENSP00000391879	1
TRIM26	ENST00000437089	?	ENSP00000395491	1
TRIM26	ENST00000454678	NM_003449.4	ENSP00000410446	1
ZNRD1	ENST00000332435	NM_170783.3	ENSP00000331111	1
ZNRD1	ENST00000359374	NM_014596.5	ENSP00000352333	1
ZNRD1	ENST00000376782	NM_001278785.1	ENSP00000365978	1
ZNRD1	ENST00000376785	NM_001278786.1	ENSP00000365981	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FLOT1	ENST00000484168	?	1
FLOT1	ENST00000484693	?	1
GNL1	ENST00000462708	?	1
GNL1	ENST00000464231	?	1
HCG19P	ENST00000448756	?	2
PAIP1P1	ENST00000446875	?	1
TRIM26	ENST00000480999	?	2
UBQLN1P1	ENST00000441056	?	1
XXbac-BPG283O16.9	ENST00000624252	?	1
ZNRD1	ENST00000463141	?	1
ZNRD1	ENST00000471008	?	1
ZNRD1-AS1	ENST00000444051	?	1
ZNRD1-AS1	ENST00000376797	?	2