Locus 253 (rs33932084)

SNiPAcard

Block annotations

Block info

genomic range	chr6:27,771,106-28,768,698
block size	997,593 bp
variant count	151 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.110 [-3.56 – 5.072]	gene(s) hit or close-by	AL009179.1, AL022393.7, AL022393.9, AL049822.1, AL121932.1, COX11P1, GPX6, HIST1H1B, HIST1H2AI, HIST1H2AJ, HIST1H2AK, HIST1H2AL, HIST1H2AM, HIST1H2BL, HIST1H2BM, HIST1H2BN, HIST1H2BO, HIST1H2BPS2, HIST1H3H, HIST1H3I, HIST1H4J, HIST1H4K, HIST1H4L, HIST1H4PS1, LINC00533, NKAPL, NOP56P1, OR1F12, OR2B2, OR2B7P, OR2B8P, OR2W2P, OR2W4P, OR2W6P, PGBD1, RNU7-26P, RP1-265C24.5, RP5-874C20.6, RP5-874C20.7, RP5-874C20.8, RPLP2P1, RPSAP2, TOB2P1, Z98744.1, Z98744.2, ZBED9, ZKSCAN3, ZKSCAN8, ZNF165, ZNF192P1, ZNF192P2, ZSCAN12, ZSCAN12P1, ZSCAN16, ZSCAN16-AS1, ZSCAN23, ZSCAN26, ZSCAN31, ZSCAN9
phastCons	μ = 0.083 [0 – 1]	eQTL gene(s)	ABT1, AL022393.7, BTN2A2, BTN3A2, BTN3A3, HIST1H2BK, HIST1H3I, HIST1H3J, HIST1H4J, HLA-C, HLA-F-AS1, HLA-G, LINC01556, OR2B7P, OR2B8P, PRSS16, RP1-265C24.5, RP1-313I6.12, ZFP57, ZKSCAN3, ZNF165, ZNF192P1, ZNF391, ZSCAN23, ZSCAN26, ZSCAN31
GERP++	μ = -0.199 [-6.81 – 4.37]	potentially regulated gene(s)	GPX5, GPX5, GPX6, GPX6, HIST1H1B, HIST1H2AI, HIST1H2BL, HIST1H2BPS2, HIST1H4J, HIST1H4K, HIST1H4L, OR2B2, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2U1P, OR2U1P, OR2U1P, OR2U1P, OR2U1P, RSL24D1P1, ZKSCAN3, ZSCAN12P1
CADD score	μ = 4.950 [0.003 – 22.8]	disease gene(s)	ZFP57

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
betaine	7.17×10-6	Metabolomics GWAS Server	24816252		24
phenol sulfate	4.67×10-6	Metabolomics GWAS Server	24816252		5
Lupus erythematosus, systemic	6.28×10-6	dbGaP	pha002867	dbGaP	4
Lupus erythematosus, systemic	1.21×10-9	dbGaP	pha002848	dbGaP	4

Disease gene annotation

gene	trait	source DB	source entry/link	
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410	OMIM®
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410	OMIM®
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886	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)
OR2B2	missense variant	ENST00000303324	NM_033057.2	ENSP00000304419	2	2			2

PGBD1	missense variant	ENST00000259883	NM_001184743.1, NM_032507.3	ENSP00000259883	S/N	aGt/aAt	?	?	1
ZSCAN12	missense variant	ENST00000396827	?	ENSP00000380039	T/M	aCg/aTg	?	?	1
ZSCAN12	missense variant	ENST00000361028	?	ENSP00000354305	T/M	aCg/aTg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
AL022393.7	?	ENSG00000216901	tibial nerve	2.04×10^{-7} (p-value)	GTEx Portal V6	111
ZFP57	?	ENSG00000204644	tibial nerve	3.03×10^{-7} (p-value)	GTEx Portal V6	39
ZSCAN26	?	ENSG00000197062	thyroid	1.92×10^{-7} (p-value)	GTEx Portal V6	143
OR2B8P	?	ENSG00000182477	thyroid	5.00×10^{-10} (p-value)	GTEx Portal V6	148
OR2B7P	?	ENSG00000187763	thyroid	5.70×10^{-7} (p-value)	GTEx Portal V6	145
ZNF192P1	?	ENSG00000226314	thyroid	3.51×10^{-7} (p-value)	GTEx Portal V6	110
RP1-265C24.5	?	ENSG00000219392	thyroid	3.29×10^{-5} (p-value)	GTEx Portal V6	14
ZFP57	?	ENSG00000204644	thyroid	2.42×10^{-11} (p-value)	GTEx Portal V6	40
RP1-265C24.5	?	ENSG00000219392	skeletal muscle	9.19×10^{-9} (p-value)	GTEx Portal V6	149
AL022393.7	?	ENSG00000216901	skeletal muscle	2.24×10^{-13} (p-value)	GTEx Portal V6	148
ZSCAN23	?	ENSG00000187987	skeletal muscle	2.13×10^{-6} (p-value)	GTEx Portal V6	110
ZNF192P1	?	ENSG00000226314	skeletal muscle	1.02×10^{-5} (p-value)	GTEx Portal V6	3
ZFP57	?	ENSG00000204644	skeletal muscle	5.24×10^{-6} (p-value)	GTEx Portal V6	4
HIST1H3J	?	ENSG00000197153	testis	7.04×10^{-7} (p-value)	GTEx Portal V6	100
AL022393.7	?	ENSG00000216901	testis	1.10×10^{-6} (p-value)	GTEx Portal V6	61
RP1-265C24.5	?	ENSG00000219392	testis	9.76×10^{-8} (p-value)	GTEx Portal V6	56
RP1-313I6.12	?	ENSG00000272009	testis	3.48×10^{-6} (p-value)	GTEx Portal V6	2
OR2B7P	?	ENSG00000187763	testis	5.52×10^{-6} (p-value)	GTEx Portal V6	2
ZSCAN26	?	ENSG00000197062	transverse colon	1.13×10^{-6} (p-value)	GTEx Portal V6	16
AL022393.7	?	ENSG00000216901	cerebellum	1.41×10^{-6} (p-value)	GTEx Portal V6	147
PRSS16	?	ENSG00000112812	cerebellum	2.09×10^{-9} (p-value)	GTEx Portal V6	64
ZNF192P1	?	ENSG00000226314	cerebellum	1.85×10^{-5} (p-value)	GTEx Portal V6	6
OR2B8P	?	ENSG00000182477	sun exposed skin	1.04×10^{-7} (p-value)	GTEx Portal V6	147
ZKSCAN3	?	ENSG00000189298	sun exposed skin	1.94×10^{-5} (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	sun exposed skin	5.44×10^{-9} (p-value)	GTEx Portal V6	40
RP1-265C24.5	?	ENSG00000219392	left ventricle	1.09×10^{-9} (p-value)	GTEx Portal V6	147
AL022393.7	?	ENSG00000216901	left ventricle	2.81×10^{-6} (p-value)	GTEx Portal V6	57
ZNF165	?	ENSG00000197279	left ventricle	1.33×10^{-5} (p-value)	GTEx Portal V6	1
ZNF192P1	?	ENSG00000226314	left ventricle	6.32×10^{-6} (p-value)	GTEx Portal V6	5
RP1-265C24.5	?	ENSG00000219392	subcutaneous adipocytes	3.87×10^{-8} (p-value)	GTEx Portal V6	113

RP1-265C24.5	?	ENSG00000219392	subcutaneous adipocytes	3.87×10-8 (p-value)	GTEx Portal V6	113
HIST1H4J	?	ENSG00000197238	subcutaneous adipocytes	2.04×10-7 (p-value)	GTEx Portal V6	142
RP1-313I6.12	?	ENSG00000272009	subcutaneous adipocytes	1.45×10-5 (p-value)	GTEx Portal V6	14
HIST1H2BK	?	ENSG00000197903	subcutaneous adipocytes	1.14×10-5 (p-value)	GTEx Portal V6	24
ZNF391	?	ENSG00000124613	subcutaneous adipocytes	1.08×10-5 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	subcutaneous adipocytes	6.15×10-7 (p-value)	GTEx Portal V6	34
ZSCAN26	ENST00000421553	ILMN_1760982	blood	1.05×10-6 (p-value)	MuTHER consortium	37
ZSCAN26	ENST00000614088					
ZSCAN26	ENST00000316606					
ZSCAN26	ENST00000619937					
ZSCAN31	ENST00000396838	ILMN_1655748	skin	7.67×10-7 (p-value)	MuTHER consortium	38
ZSCAN31	ENST00000344279		adipocyte	2.79×10-5 (p-value)	MuTHER consortium	38
ZSCAN31	ENST00000414429					
ZSCAN31	ENST00000439158					
ZSCAN31	ENST00000611469					
ZSCAN31	ENST00000446474					
HIST1H3I	ENST00000616365	ILMN_2207865	b-cell	2.79×10-4 (p-value)	Fairfax et al.	6
BTN2A2	ENST00000471116	ILMN_2256894	b-cell	4.07×10-6 (p-value)	Fairfax et al.	14
BTN2A2	ENST00000356709					
BTN2A2	ENST00000494184					
BTN2A2	ENST00000482636					
BTN2A2	ENST00000469230					
BTN2A2	ENST00000416795					
BTN2A2	ENST00000432533					
BTN2A2	ENST00000493275					
ABT1	ENST00000274849	ILMN_1658083	monocyte	4.18×10-5 (p-value)	Fairfax et al.	7
ZSCAN26	?	ENSG00000197062	transformed fibroblasts	1.09×10-5 (p-value)	GTEx Portal V6	116
ZNF192P1	?	ENSG00000226314	transformed fibroblasts	3.66×10-5 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	transformed fibroblasts	2.25×10-6 (p-value)	GTEx Portal V6	33
ZSCAN26	?	ENSG00000197062	adrenal gland	6.33×10-7 (p-value)	GTEx Portal V6	92
HIST1H4J	?	ENSG00000197238	adrenal gland	1.43×10-5 (p-value)	GTEx Portal V6	1
AL022393.7	?	ENSG00000216901	muscularis mucosae	1.61×10-5 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	muscularis mucosae	2.02×10-7 (p-value)	GTEx Portal V6	39
LINC01556	?	ENSG00000204709	aorta	1.17×10-5 (p-value)	GTEx Portal V6	36
ZFP57	?	ENSG00000204644	aorta	8.39×10-8 (p-value)	GTEx Portal V6	39
ZSCAN23	?	ENSG00000187987	tibial artery	3.47×10-5 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	tibial artery	1.37×10-7 (p-value)	GTEx Portal V6	38
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	2.75×10-52 (p-value)	Zeller et al.	3
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	1.60×10-31 (p-value)	Zeller et al.	3
?	?	ILMN_1883997	monocyte	1.36×10-32 (p-value)	Zeller et al.	3
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	2.62×10-37 (p-value)	Zeller et al.	3
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_1679389	monocyte	1.99×10-14 (p-value)	Zeller et al.	2
ZFP57	?	ENSG00000204644	pancreas	7.02×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	lung	3.76×10-8 (p-value)	GTEx Portal V6	40
ZFP57	?	ENSG00000204644	blood	9.66×10-10 (p-value)	GTEx Portal V6	40
HLA-F-AS1	?	ENSG00000214922	blood	3.22×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	breast	5.76×10-7 (p-value)	GTEx Portal V6	34
ZFP57	?	ENSG00000204644	unexposed skin	4.21×10-6 (p-value)	GTEx Portal V6	33
ZFP57	?	ENSG00000204644	stomach	5.19×10-7 (p-value)	GTEx Portal V6	34
ZFP57	?	ENSG00000204644	esophagus mucosa	6.01×10-13 (p-value)	GTEx Portal V6	40
ZSCAN26	?	ENSG00000197062	esophagus mucosa	9.75×10-6 (p-value)	GTEx Portal V6	29
ZFP57	?	ENSG00000204644	atrial appendage	3.72×10-7 (p-value)	GTEx Portal V6	33
ZFP57	?	ENSG00000204644	visceral adipocytes	7.12×10-6 (p-value)	GTEx Portal V6	27
ZFP57	?	ENSG00000204644	liver	3.48×10-6 (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
BTN3A2	?	ENSG00000186470	chr6	blood	1.67×10-7 (q-value)	SeeQTL DB (HapMap)	15
BTN3A2	ENST00000524682	209846_s_at	chr6	blood	6.40×10-16 (p-value)	Dixon et al.	4
BTN3A2	ENST00000604202						
BTN3A2	ENST00000396934						
BTN3A2	ENST00000532294						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000377708						
BTN3A2	ENST00000527422						
BTN3A2	ENST00000531055						
BTN3A2	ENST00000396948						
BTN3A2	ENST00000356386						
BTN3A2	ENST00000396934	212613_at	chr6	blood	1.40×10-13 (p-value)	Dixon et al.	4
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000244519	204820_s_at	chr6	blood	3.00×10-15 (p-value)	Dixon et al.	4
BTN3A2	ENST00000396934						
BTN3A2	ENST00000508906						

BTN3A2	ENST00000356386						
BTN3A3	ENST00000480110						
?	?	ILMN_1679389	chr6	monocyte	1.50×10-12 (p-value)	Zeller et al.	1
?	?	ILMN_1671054	chr6	monocyte	3.41×10-37 (p-value)	Zeller et al.	3
?	?	ILMN_1666078	chr6	monocyte	4.02×10-28 (p-value)	Zeller et al.	3
?	?	ILMN_1752592	chr6	monocyte	5.77×10-19 (p-value)	Zeller et al.	3
?	?	ILMN_1715169	chr6	monocyte	3.28×10-12 (p-value)	Zeller et al.	1
HLA-G	ENST00000376828	ILMN_1656670	chr6	monocyte	1.65×10-14 (p-value)	Zeller et al.	2
HLA-G	ENST00000360323						
HLA-G	ENST00000478519						
HLA-G	ENST00000478355						
HLA-G	ENST00000428701						
HLA-G	ENST00000376815						
HLA-G	ENST00000376818						
HLA-C	ENST00000470363	ILMN_2150787	chr6	monocyte	5.78×10-14 (p-value)	Zeller et al.	1
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
E2F4	MA0541.1	3	no	0.000	1
MEF2A	MA0558.1	21	no	0.000	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000047721		
ENCP00000047738		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000443358	1	ENCP00000047719	HIST1H2BPS2
		ENCP00000047723	HIST1H4L
		ENCP00000047717	HIST1H4K
		ENCP00000047716	HIST1H4J
		ENCP00000047721	HIST1H1B
ENCE00000443289	1	ENCP00000047711	RSL24D1P1
ENCE00000443591	1	ENCP00000047762	GPX6 GPX6
		ENCP00000047757	ZKSCAN3
ENCE00000443338	1	ENCP00000047713	HIST1H2AI HIST1H2BL
ENCE00000443415	1	ENCP00000047727	OR2B2
ENCE00000443677	1	ENCP00000047794	OR2U1P OR2U1P

			OR2U1P OR2B4P OR2B4P OR2B4P OR2B4P OR2U1P OR2B4P OR2B4P OR2B4P OR2U1P
ENCE00000443536	1	ENCP00000047763	GPX5 GPX5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001702839 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, SP1, H4K5ac, H3K27me3, CTCF, PolII, Jund, TAF7, Nrsf, H3K27ac, TAF1, H3K4me2
		HSMMtube	H2AZ, H3K4me2, DNase1
		blood (K562)	TAF1, Cfos, PolII, GTF2B, H4K20me1, H3K4me2, H3K36me3, H3K4me3, Ini1, Brg1, DNase1, H3K27ac, Max, TAF7, BCLAF1, CTCF, MEF2A, H3K9ac, Cmyc, H3K79me2, HEY1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, DNase1
		liver (HepG2)	H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1, H3K4me2, H2AZ, PolII, TAF1, H3K4me1
		blood (GM12878)	POU2F2, Cmyc, PolII, BCLAF1, H2AZ, DNase1, Yy1, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	CTCF, H3K27ac, H3K9ac, PolII, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	Ini1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	Cjun, Max, H3K4me3, H3K4me2, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00001214443 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K36me3, H3K27me3, PolII, Jund, TAF7, p300, Nrsf, Sin3Ak20, H3K27ac, Yy1, RXRA, Tcf12, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3
		HSMMtube	DNase1, H2AZ
		blood (K562)	H3K4me2, H3K36me3, DNase1, H3K4me3, Ini1, Brg1, H3K27ac, Max, BCLAF1, H3K9ac, H3K79me2, HEY1, TAF1, PolII
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		liver (HepG2)	DNase1, H3K27me3, H3K4me3, Gabp, Jund, PolII, TAF1, ELF1, p300, ZBTB33, H3K4me2, H3K9ac, H3K27ac
		lung (IMR90)	H3K36me3, H3K4me3, H3K9ac, H3K4me2, DNase1
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, ELF1, POU2F2, TAF1, PolII, BCLAF1, Tcf12, DNase1, Yy1
		nervous (NH-A)	H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, H3K4me1
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K36me3, DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K4me3, H3K4me2, H3K9ac, PolII, DNase1, Cmyc
		A549	H3K27ac, DNase1, H3K9ac, H3K4me3
ENSR00001214446 (promoter)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K9ac, H4K5ac, H3K27me3, CTCF, PolII, Jund, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, SP1
		HSMMtube	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		blood (K562)	Cfos, Cmyc, PolII, H3K4me2, H3K36me3, H3K4me3, Cjun, TAF1, Ini1, Brg1, DNase1, Egr1, H3K27ac, Max, SP1, H3K9ac, H3K79me2, HEY1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	PolII, H3K79me2, DNase1, TAF1, H3K4me1, H3K4me2, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3
		blood (GM12878)	PolII, DNase1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	H3K9ac, DNase1, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac

		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K27ac, H3K9ac, PolII, H3K4me2, H3K4me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K4me3, Gabp, Cfos, PolII, Cmyc, H3K36me3, Max, TAF1, Ini1, H3K9ac, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K27ac, PolII, DNase1, Cmyc, H3K9ac, Cjun, Max, H3K4me3, H3K4me2
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001494372 (enhancer)	1	endothelium (HUVEC)	H3K27me3
		blood (K562)	PolII, H3K36me3
ENSR00001702845 (CTCF binding site)	1	endothelium (HUVEC)	H3K27me3
		blood (K562)	PolII, H3K36me3
ENSR00001214452 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, SP1, H3K9ac, H3K4me3, H4K5ac, H3K36me3, CTCF, PolII, Rad21, Egr1, Jund, TAF7, Sin3Ak20, H3K27ac, Yy1, BCL11A, TAF1, H3K4me2
		HSMMtube	DNase1, H2AZ, H3K4me2
		blood (K562)	H3K9ac, HEY1, TAF1, Jund, H3K4me3, PolII, GTF2B, H3K4me2, BCLAF1, TAF7, Ini1, Brg1, H3K27ac
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H3K9ac, DNase1, H2AZ, H3K4me3, H3K27ac, H3K4me2
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K9ac, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4me2, DNase1, H3K27ac
		blood (GM12878)	POU2F2, Cmyc, PU1, PolII, BCLAF1, H2AZ, DNase1, Yy1, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac, Cjun
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K27ac, H3K9ac, PolII
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me2, H3K9ac, DNase1, H3K27ac, H3K4me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K9ac, PolII, DNase1, H3K4me2, H3K4me3, Cjun
		A549	H3K4me3, DNase1
ENSR00001214455 (promoter)	1	embryonic stem cell (H1ESC)	H3K9ac, H3K4me3, SP1, H3K4me2, H4K5ac, H3K36me3, H3K27me3, CTCF, PolII, Jund, TAF7, p300, H3K27ac, Yy1, Tcf12, TAF1
		HSMMtube	DNase1, H2AZ, H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		blood (GM12878)	H3K9ac, POU2F2, PolII, BATF, H2AZ, DNase1, ELF1, H3K4me3, H3K27ac, Cfos, H3K4me2, CTCF
		lung (IMR90)	H3K4me2, H4K5ac, H3K4me3, H3K4ac, H2BK120ac, H4K91ac, H3K56ac, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	CTCF, DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K9ac, H3K27ac, H3K4me3, DNase1
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2, CTCF
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27ac, H3K9ac, H3K4me3, DNase1
		endothelium (HUVEC)	Max, DNase1, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, Cmyc
		A549	DNase1, H3K9ac, H3K4me3, H3K27ac
ENSR00001214477 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Gabp, TAF7, CTCF, Yy1, TAF1, H3K4me2, H3K4me3
		HSMMtube	DNase1, H2AZ, H3K4me2
		blood (K562)	H3K27ac, Max, H3K9ac, Gabp, HEY1, H2AZ, TAF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me3, DNase1, CTCF, H3K9ac, Gabp, PolII, TAF1, H2AZ, H3K4me2
		blood (GM12878)	TAF1, PolII, BCLAF1, H2AZ, DNase1, Gabp, ELF1, H3K4me3, H3K27ac, CTCF, H3K9ac
		lung (IMR90)	H4K5ac, H3K4me3, CTCF, H3K4me2
		nervous (NH-A)	H3K4me3, H3K4me2, H3K9ac, DNase1
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ

		breast (HMEC)	H3K4me2, CTCF, H3K4me3
		cervix (HeLa-S3)	H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF, DNase1, H3K9ac, Ini1
		monocytes (Monocytes-CD14+)	H3K4me3, H3K9ac, CTCF
		endothelium (HUVEC)	H3K4me3, H3K4me2, PolII, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001214498 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, TAF7, Sin3Ak20, H3K9ac, Yy1, TAF1, H3K4me2, H3K4me3, Gabp, Egr1, H3K36me3, H3K27me3, CTCF, PolII, Rad21
		HSMMtube	DNase1, H3K36me3, H3K4me3, H3K4me2
		blood (K562)	Yy1, Egr1, H3K27ac, Max, SP2, H3K9ac, Gabp, Cmyc, H3K79me2, H2AZ, TAF1, ELF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, Gabp, Yy1, PolII, TAF1, ELF1, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K36me3, H3K27me3
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, H3K9ac, DNase1
		blood (GM12878)	TAF1, PolII, BCLAF1, H2AZ, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K9ac
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K36me3
		Osteobl	H2AZ, H3K4me3, H3K36me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, TAF1, PolII, H3K4me3, Gabp, H3K36me3, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K9ac, PolII, DNase1, Cmyc, H3K4me3, H3K4me2, Max
		A549	DNase1, H3K4me3
ENSR00001214514 (enhancer)	1	liver (HepG2)	H3K4me1, H3K36me3
ENSR00001494388 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, SP1, Srf, H3K9ac, H3K4me3, PolII, TAF7
		HSMMtube	H3K4me3, H3K27ac, H3K4me2, H3K9ac, H2AZ, DNase1
		blood (K562)	Ini1, Brg1, Gata2, Egr1, H3K4me3, H3K27ac, PU1, ZBTB7A, H3K9ac, Gabp, HEY1, H2AZ, TAF1, Nrsf, Cjun, Cfos, PolII, H3K4me2, H3K4me1, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H2AZ, H3K4me3, H3K27ac
		liver (HepG2)	FOSL2, Gabp, Jund, PolII, TAF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K9ac, H4K5ac, H3K4me2, H3K27ac, DNase1, H3K18ac
		blood (GM12878)	PolII, Srf, H2AZ, DNase1, Yy1, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Gabp, PolII, TAF1, H3K4me3, H3K9ac, Jund, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, H3K4me3, H3K4me2, H3K9ac, PolII, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
ENSR00001214525 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	DNase1
		A549	H3K27me3
ENSR00001214530 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, PolII, Rad21, Sin3Ak20, CTCF, H3K27ac, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K9ac, CTCF, H3K4me2, DNase1, H3K36me3, H2AZ, H3K27ac
		blood (K562)	TAF1, ELF1, PolII, H3K4me2, CTCF, H3K4me1, H3K36me3, DNase1, H3K4me3, Rad21, H2AZ, H3K27me3, Ini1, Brg1, H3K27ac, Max, SP2, Nfe2, H3K9ac, Gabp, H3K79me2, HEY1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	CTCF, H3K4me3, H3K27me3, DNase1, H3K27ac, H3K9ac, Gabp, PolII, Rad21, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2
		blood (GM12878)	CTCF, H3K9ac, H3K4me2, H3K27ac, DNase1, PolII, H2AZ, Yy1, Gabp, ELF1, Rad21, H3K79me2, H3K4me3
		lung (IMR90)	DNase1, H3K4me2, H3K36me3, H3K4me3, H3K9ac, CTCF
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF
		NHLF	DNase1, H3K4me3, CTCF, H3K27ac

		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, CTCF
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, PolII, H3K4me3, Gabp, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, CTCF, PolII, H3K4me3, H3K4me2, Cjun, DNase1
		A549	H3K36me3, DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00001214540 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, Egr1, Gabp, TAF7, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, PolII, H3K27me3, H3K36me3
		HSMMtube	CTCF, DNase1, H3K36me3, H2AZ, H3K4me3, H3K4me2, H3K9ac
		blood (K562)	Ini1, Brg1, Egr1, H3K27ac, Max, SP2, BCLAF1, H3K4me3, DNase1, ZBTB7A, H3K9ac, Gabp, ETS1, Cmyc, H3K79me2, HEY1, H2AZ, TAF1, ELF1, CTCFL, PolII, H3K4me2, CTCF, H3K36me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K36me3
		liver (HepG2)	DNase1, H3K27me3, H3K36me3, H3K4me3, Cmyc, Gabp, PolII, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF
		blood (GM12878)	ELF1, H3K79me2, H3K4me3, H3K27ac, CTCF, H3K9ac, H3K36me3, Gabp, Yy1, TAF1, PolII, BCLAF1, Srf, H2AZ, ETS1, DNase1
		lung (IMR90)	H4K8ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac, CTCF, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K36me3, H3K4me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, H3K36me3
		NHLF	H3K36me3, DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me3, H3K36me3, H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac
		breast (HMEC)	CTCF, H3K4me2, H3K4me3, H3K9ac
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K9ac, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, PolII, CTCF, H3K27me3, H3K36me3
		A549	H3K36me3, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00001494403 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K27me3, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, DNase1, H3K27me3, H2AZ, H3K4me3
		blood (K562)	PolII, Ini1, DNase1, PolIII, H3K27ac, Max, H3K9ac, Cmyc, H2AZ, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me2, DNase1
		liver (HepG2)	H2AZ, H3K4me2, H3K4me3, H3K27me3
		blood (GM12878)	H3K4me2, H3K4me3, H2AZ
		lung (IMR90)	H3K4me3, H3K4me2
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K4me2, H3K4me3, H3K27me3
		NHLF	H3K4me3
		Osteobl	H3K27me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K27me3, H3K4me3, H3K4me2
		breast (HMEC)	H3K4me2, H3K4me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3, H3K4me2, H3K4me3
		A549	H3K9ac, H3K4me2, H3K4me3
ENSR00000487476 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3, Egr1, H3K4me2, H3K4me3
		HSMMtube	H3K27me3, DNase1
		blood (K562)	PolII, Ini1, DNase1, PolIII, Egr1, Max, H2AZ, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me3, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3, H3K4me2, DNase1
		skin (NHEK)	DNase1, H3K4me2
ENSR00001702885 (CTCF binding site)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K27me3, Egr1, H3K4me3, H3K4me2
		HSMMtube	H3K27me3, DNase1

		blood (K562)	PolII, Ini1, DNase1, PolIII, Egr1, Max, H2AZ, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me3, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	DNase1, H3K4me2, H3K4me3
		skin (NHEK)	H3K4me2, DNase1
ENSR00000264184 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K27me3, H3K9ac, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	Ini1, Brg1, DNase1, H3K27ac, Max, H3K9ac, H2AZ, PolII, Cmyc, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me3
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, H3K4me3, Max, Cmyc, H3K9ac, PolII
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K4me1, H2AZ, H3K4me2, H3K4me3, H3K27me3, DNase1
		A549	DNase1, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, H3K4me2, H3K4me3
ENSR00001702887 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3, H3K36me3
		blood (K562)	H3K4me2, H2AZ
ENSR00000487478 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K4me2, H3K27me3, H3K4me3, DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		skin (NHEK)	H3K27me3
		muscle (HSMM)	H3K27me3
		breast (HMEC)	H3K27me3
ENSR00000264195 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K4me2, H3K27me3, H3K4me3, DNase1
		HSMMtube	H3K27me3
		blood (K562)	PolII, Ini1, Brg1, DNase1, PolIII, H3K27ac, Max, H3K9ac, H2AZ, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3
		muscle (HSMM)	DNase1
		liver (HepG2)	H3K4me3, H3K4me2, H3K27me3
		blood (GM12878)	H3K4me3
		lung (IMR90)	H3K4me3
		nervous (NH-A)	H3K27me3, H3K4me2
		skin (NHEK)	DNase1, H3K4me3, H3K27me3
		Osteobl	H3K27me3, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me2
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K4me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001494411 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, H3K4me2
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K27me3
		blood (K562)	PU1, MEF2A, H3K4me2, H3K4me3, Max, PolIII, PolII, Ini1, DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, H3K4me2
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me2
		nervous (NH-A)	H3K27me3
		A549	DNase1, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL009179.1	downstream gene variant	1163	ENST00000625569	?	?	1
AL022393.7	upstream gene variant, downstream gene variant	380	ENST00000405613	?	?	2

	gene variant					
AL022393.9	upstream gene variant, downstream gene variant	1124	ENST00000625142	?	?	3
AL049822.1	downstream gene variant	1626	ENST00000631369	?	?	1
AL121932.1	upstream gene variant	4597	ENST00000577570	?	?	1
COX11P1	upstream gene variant	2809	ENST00000489116	?	?	1
HIST1H1B	downstream gene variant, upstream gene variant	488	ENST00000331442	NM_005322.2	ENSP00000330074	3
HIST1H2AI	upstream gene variant	949	ENST00000358739	NM_003509.2	ENSP00000351589	2
HIST1H2AJ	upstream gene variant	1423	ENST00000333151	NM_021066.2	ENSP00000328484	1
HIST1H2AK	downstream gene variant, upstream gene variant	470	ENST00000618958	NM_003510.2	ENSP00000482431	3
HIST1H2AL	downstream gene variant	560	ENST00000613174	NM_003511.2	ENSP00000482538	4
HIST1H2AM	upstream gene variant	4975	ENST00000359611	NM_003514.2	ENSP00000352627	1
HIST1H2BL	downstream gene variant	194	ENST00000377401	NM_003519.3	ENSP00000366618	2
HIST1H2BM	downstream gene variant	739	ENST00000621112	NM_003521.2	ENSP00000477907	1
HIST1H2BN	upstream gene variant	1068	ENST00000449538	?	ENSP00000446031	1
HIST1H2BN	upstream gene variant, downstream gene variant	509	ENST00000396980	?	ENSP00000380177	5
HIST1H2BN	upstream gene variant	1124	ENST00000606613	?	ENSP00000475942	1
HIST1H2BN	upstream gene variant, downstream gene variant	283	ENST00000612898	NM_003520.3	ENSP00000483903	2
HIST1H2BO	downstream gene variant	4281	ENST00000616182	NM_003527.4	ENSP00000477527	1
HIST1H2BPS2	upstream gene variant	1906	ENST00000402707	?	?	3
HIST1H3H	upstream gene variant, downstream gene variant	2814	ENST00000369163	NM_003536.2	ENSP00000358160	2
HIST1H3I	downstream gene variant, upstream gene variant	833	ENST00000616365	NM_003533.2	ENSP00000483283	3
HIST1H4J	downstream gene variant	1734	ENST00000355057	NM_021968.3	ENSP00000347168	1
HIST1H4K	downstream gene variant	107	ENST00000611927	NM_003541.2	ENSP00000479794	1
HIST1H4L	downstream gene variant	51	ENST00000618305	NM_003546.2	ENSP00000480960	2
HIST1H4PS1	upstream gene variant	3747	ENST00000406085	?	?	1
LINC00533	upstream gene variant	2510	ENST00000456103	?	?	1
NKAPL	upstream gene variant	1787	ENST00000343684	NM_001007531.2	ENSP00000345716	1
NOP56P1	downstream gene variant, upstream gene variant	3129	ENST00000440030	?	?	3
OR1F12	upstream gene variant	2165	ENST00000402365	?	?	1
OR2B2	upstream gene variant	3838	ENST00000303324	NM_033057.2	ENSP00000304419	1
OR2B7P	downstream gene variant	2105	ENST00000429302	?	?	1
OR2B8P	downstream gene variant	3756	ENST00000432841	?	?	1
OR2W2P	downstream gene variant	595	ENST00000406313	?	?	1
OR2W4P	upstream gene variant	1559	ENST00000406496	?	?	1
PGBD1	downstream gene variant	1647	ENST00000259883	NM_001184743.1, NM_032507.3	ENSP00000259883	1
RNU7-26P	upstream gene variant	558	ENST00000458980	?	?	2
RP1-265C24.5	upstream gene variant	1648	ENST00000405929	?	?	4
RP5-874C20.6	downstream gene variant, upstream	1452	ENST00000605757	?	?	3

	gene variant					
RP5-874C20.7	downstream gene variant, upstream gene variant	1496	ENST00000615674	?	?	4
RP5-874C20.8	downstream gene variant	822	ENST00000621053	?	ENSP00000481142	2
RPLP2P1	upstream gene variant	652	ENST00000398220	?	?	1
RPSAP2	upstream gene variant, downstream gene variant	811	ENST00000416785	?	?	3
TOB2P1	upstream gene variant	1220	ENST00000469761	?	?	2
Z98744.1	upstream gene variant, downstream gene variant	480	ENST00000629628	?	?	3
Z98744.2	upstream gene variant	569	ENST00000627472	?	?	4
ZKSCAN3	upstream gene variant	2847	ENST00000377255	NM_001242894.1	ENSP00000366465	1
ZKSCAN3	upstream gene variant	2847	ENST00000341464	NM_001242895.1	ENSP00000341883	1
ZKSCAN3	upstream gene variant	2811	ENST00000252211	NM_024493.3	ENSP00000252211	1
ZKSCAN8	downstream gene variant	3257	ENST00000536028	?	ENSP00000439117	1
ZKSCAN8	downstream gene variant	2263	ENST00000457389	NM_001278119.1	ENSP00000402948	1
ZKSCAN8	downstream gene variant	1981	ENST00000330236	NM_001278122.1, NM_006298.3, NM_001278121.1	ENSP00000332750	4
ZKSCAN8	downstream gene variant	2225	ENST00000606198	?	ENSP00000475589	1
ZNF165	downstream gene variant	1608	ENST00000377325	NM_003447.3	ENSP00000366542	2
ZNF192P1	upstream gene variant, downstream gene variant	116	ENST00000565888	?	?	6
ZNF192P1	upstream gene variant, downstream gene variant	312	ENST00000562227	?	?	5
ZNF192P1	upstream gene variant, downstream gene variant	127	ENST00000440790	?	?	4
ZNF192P1	upstream gene variant, downstream gene variant	338	ENST00000570126	?	?	4
ZNF192P1	upstream gene variant, downstream gene variant	107	ENST00000570042	?	?	4
ZNF192P2	upstream gene variant	4950	ENST00000402215	?	?	1
ZSCAN12	downstream gene variant, upstream gene variant	1000	ENST00000396827	?	ENSP00000380039	2
ZSCAN12	downstream gene variant, upstream gene variant	997	ENST00000361028	?	ENSP00000354305	2
ZSCAN12P1	downstream gene variant	4359	ENST00000529104	?	?	1
ZSCAN12P1	upstream gene variant	1256	ENST00000406489	?	?	1
ZSCAN16	upstream gene variant	758	ENST00000340487	NM_025231.1	ENSP00000366527	2
ZSCAN16-AS1	downstream gene variant	95	ENST00000602810	?	?	1
ZSCAN16-AS1	downstream gene variant	92	ENST00000600652	?	?	1
ZSCAN23	upstream gene variant	662	ENST00000486481	?	?	1
ZSCAN23	upstream gene variant	684	ENST00000481983	?	ENSP00000435430	1
ZSCAN23	upstream gene variant	697	ENST00000289788	NM_001012455.1	ENSP00000289788	1
ZSCAN26	downstream gene variant	4201	ENST00000316606	NM_152736.5	ENSP00000484931	2
ZSCAN26	downstream gene variant	4178	ENST00000619937	?	ENSP00000484018	2
ZSCAN26	downstream gene variant	4181	ENST00000614088	NM_001287421.1	ENSP00000479314	2
ZSCAN26	downstream gene variant	4872	ENST00000623276	NM_001111039.2	ENSP00000485228	1
ZSCAN26	downstream gene variant	4181	ENST00000421553	NM_001023560.3	ENSP00000481707	2

ZSCAN31	downstream gene variant, upstream gene variant	473	ENST00000414431 ?	ENSP00000407529	5
ZSCAN31	upstream gene variant	1721	ENST00000439628 ?	ENSP00000392552	2
ZSCAN31	downstream gene variant, upstream gene variant	671	ENST00000447021 ?	ENSP00000416108	5
ZSCAN31	downstream gene variant, upstream gene variant	293	ENST00000476001 ?	?	3
ZSCAN31	downstream gene variant, upstream gene variant	442	ENST00000344279 NM_030899.4, NM_001243241.1	ENSP00000345339	5
ZSCAN31	upstream gene variant	1721	ENST00000434036 ?	ENSP00000416225	2
ZSCAN31	downstream gene variant, upstream gene variant	330	ENST00000414429 ?	ENSP00000390076	3
ZSCAN31	downstream gene variant, upstream gene variant	232	ENST00000435857 ?	ENSP00000391235	5
ZSCAN31	downstream gene variant	1823	ENST00000481934 ?	?	2
ZSCAN31	downstream gene variant, upstream gene variant	232	ENST00000611469 NM_001243244.1, NM_001243242.1	ENSP00000480254	5
ZSCAN31	downstream gene variant, upstream gene variant	473	ENST00000446474 NM_001243243.1	ENSP00000402937	5
ZSCAN31	downstream gene variant, upstream gene variant	1730	ENST00000396838 ?	ENSP00000380050	4
ZSCAN31	downstream gene variant, upstream gene variant	1721	ENST00000426434 ?	ENSP00000398680	4
ZSCAN31	upstream gene variant	1860	ENST00000444081 ?	ENSP00000415040	2
ZSCAN31	downstream gene variant, upstream gene variant	1156	ENST00000439636 ?	ENSP00000412519	5
ZSCAN31	upstream gene variant	1739	ENST00000446222 ?	ENSP00000411033	2
ZSCAN31	downstream gene variant, upstream gene variant	473	ENST00000439158 NM_001135216.1	ENSP00000413705	5
ZSCAN31	downstream gene variant, upstream gene variant	409	ENST00000453745 ?	ENSP00000389479	5
ZSCAN9	upstream gene variant, downstream gene variant	738	ENST00000527844 ?	ENSP00000436166	4
ZSCAN9	upstream gene variant, downstream gene variant	1167	ENST00000526391 ?	ENSP00000476254	4
ZSCAN9	upstream gene variant, downstream gene variant	271	ENST00000531979 NM_001199480.1	ENSP00000433402	3
ZSCAN9	upstream gene variant, downstream gene variant	15	ENST00000527436 ?	ENSP00000433468	3
ZSCAN9	upstream gene variant, downstream gene variant	287	ENST00000252207 NM_006299.4	ENSP00000252207	3
ZSCAN9	upstream gene variant	1376	ENST00000531941 ?	ENSP00000433307	1
ZSCAN9	upstream gene variant	1754	ENST00000531981 ?	ENSP00000475719	1
ZSCAN9	upstream gene variant, downstream gene variant	287	ENST00000425468 NM_001199479.1	ENSP00000404074	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
HIST1H1B	ENST00000331442	NM_005322.2	ENSP00000330074	G	ggT/ggC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPX6	ENST00000612264	?	ENSP00000483462	1
GPX6	ENST00000361902	NM_182701.1	ENSP00000354581	1
GPX6	ENST00000474923	?	ENSP00000417364	1
GPX6	ENST00000483058	?	?	1
HIST1H2BN	ENST00000606613	?	ENSP00000475942	5

HIST1H2BN	ENST00000449538	?	ENSP00000446031	5
HIST1H2BN	ENST00000396980	?	ENSP00000380177	5
PGBD1	ENST00000259883	NM_001184743.1, NM_032507.3	ENSP00000259883	6
RP5-874C20.8	ENST00000621053	?	ENSP00000481142	1
ZBED9	ENST00000530247	?	?	1
ZKSCAN3	ENST00000377255	NM_001242894.1	ENSP00000366465	4
ZKSCAN3	ENST00000252211	NM_024493.3	ENSP00000252211	4
ZKSCAN3	ENST00000341464	NM_001242895.1	ENSP00000341883	4
ZKSCAN8	ENST00000536028	?	ENSP00000439117	2
ZKSCAN8	ENST00000330236	NM_001278122.1, NM_006298.3, NM_001278121.1	ENSP00000332750	2
ZKSCAN8	ENST00000457389	NM_001278119.1	ENSP00000402948	2
ZKSCAN8	ENST00000606198	?	ENSP00000475589	2
ZNF192P1	ENST00000562227	?	?	2
ZNF192P1	ENST00000570126	?	?	1
ZNF192P1	ENST00000440790	?	?	3
ZNF192P1	ENST00000570042	?	?	2
ZSCAN12	ENST00000396827	?	ENSP00000380039	8
ZSCAN12	ENST00000361028	?	ENSP00000354305	8
ZSCAN16-AS1	ENST00000600652	?	?	1
ZSCAN16-AS1	ENST00000602810	?	?	1
ZSCAN26	ENST00000617168	?	ENSP00000481024	1
ZSCAN26	ENST00000316606	NM_152736.5	ENSP00000484931	1
ZSCAN26	ENST00000614088	NM_001287421.1	ENSP00000479314	1
ZSCAN26	ENST00000421553	NM_001023560.3	ENSP00000481707	1
ZSCAN26	ENST00000611552	NM_001287422.1	ENSP00000478769	1
ZSCAN26	ENST00000619937	?	ENSP00000484018	1
ZSCAN26	ENST00000623276	NM_001111039.2	ENSP00000485228	1
ZSCAN31	ENST00000426756	?	ENSP00000406376	3
ZSCAN31	ENST00000446222	?	ENSP00000411033	5
ZSCAN31	ENST00000414429	?	ENSP00000390076	6
ZSCAN31	ENST00000396838	?	ENSP00000380050	5
ZSCAN31	ENST00000426434	?	ENSP00000398680	5
ZSCAN31	ENST00000439628	?	ENSP00000392552	5
ZSCAN31	ENST00000476001	?	?	4
ZSCAN31	ENST00000434036	?	ENSP00000416225	5
ZSCAN31	ENST00000444081	?	ENSP00000415040	5
ZSCAN9	ENST00000531941	?	ENSP00000433307	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
HIST1H2BN	ENST00000612898	NM_003520.3	ENSP00000483903	1
ZKSCAN8	ENST00000330236	NM_001278122.1, NM_006298.3, NM_001278121.1	ENSP00000332750	1

ZSCAN12	ENST00000361028	?	ENSP00000354305	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZSCAN9	ENST00000531979	NM_001199480.1	ENSP00000433402	1
ZSCAN9	ENST00000425468	NM_001199479.1	ENSP00000404074	1
ZSCAN9	ENST00000531981	?	ENSP00000475719	1
ZSCAN9	ENST00000252207	NM_006299.4	ENSP00000252207	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AL022393.9	ENST00000625142	?	1
HIST1H4PS1	ENST00000406085	?	1
OR2W4P	ENST00000406496	?	1
OR2W6P	ENST00000439689	?	1
ZNF192P1	ENST00000562227	?	1
ZNF192P1	ENST00000570126	?	1
ZNF192P1	ENST00000440790	?	1
ZNF192P1	ENST00000570042	?	1
ZSCAN12P1	ENST00000406489	?	1
ZSCAN12P1	ENST00000529104	?	2