Locus 95 (rs11715835)

SNiPAcard

Block annotations

Block info

genomic range	chr3:48,688,114-49,269,270
block size	581,157 bp
variant count	107 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.089 [-2.931 – 4.247]	**gene(s) hit or close-by**	AC141002.1, ARIH2, ARIH2OS, C3orf84, CCDC36, CCDC71, CELSR3, DALRD3, IMPDH2, IP6K2, KLHDC8B, LAMB2, MIR6890, NCKIPSD, NDUFAF3, P4HTM, PRKAR2A, PRKAR2A-AS1, QARS, QRICH1, RN7SL182P, RP11-694I15.7, RP13-131K19.1, RP13-131K19.2, RP13-131K19.6, SLC25A20, USP19, WDR6
phastCons	μ = 0.113 [0 – 1]	**eQTL gene(s)**	AMT, ARIH2, C3orf62, CCDC36, CCDC71, CELSR3, IP6K2, KLHDC8B, MIR5193, NCKIPSD, NICN1, RBM6, RP11-3B7.1, SEMA3F, SLC26A6, TMA7, TMEM89, UBA7, USP4, WDR6
GERP++	μ = -0.294 [-10 – 4.95]	**potentially regulated gene(s)**	AMT, AMT, ARIH2, ATRIP, CCDC36, CELSR3, COL7A1, COL7A1, DALRD3, IMPDH2, IP6K2, KLHDC8B, NCKIPSD, P4HTM, PRKAR2A, QARS, SLC26A6, SPINK8, UCN2, USP19, WDR6
CADD score	μ = 5.047 [0.048 – 21.7]	**disease gene(s)**	KLHDC8B, AMT, SLC25A20, COL7A1, NDUFAF3, QARS, LAMB2, ATRIP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
KLHDC8B	LYMPHOMA, HODGKIN, CLASSIC	OMIM	MIM:236000
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OMIM	MIM:212138
COL7A1	NAIL DISORDER, NONSYNDROMIC CONGENITAL, 8	OMIM	MIM:607523
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, PRETIBIAL	OMIM	MIM:131850
COL7A1	EPIDERMOLYSIS BULLOSA PRURIGINOSA	OMIM	MIM:604129
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL DOMINANT	OMIM	MIM:131750
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE	OMIM	MIM:226600
COL7A1	EPIDERMOLYSIS BULLOSA WITH CONGENITAL LOCALIZED ABSENCE OF SKIN [...]	OMIM	MIM:132000
COL7A1	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN	OMIM	MIM:131705
NDUFAF3	MITOCHONDRIAL COMPLEX I DEFICIENCY	OMIM	MIM:252010
QARS	MICROCEPHALY, PROGRESSIVE, WITH SEIZURES AND CEREBRAL AND [...]	OMIM	MIM:615760
LAMB2	NEPHROTIC SYNDROME, TYPE 5, WITH OR WITHOUT OCULAR [...]	OMIM	MIM:614199
LAMB2	PIERSON SYNDROME	OMIM	MIM:609049
ATRIP	Seckel syndrome	OrphaNet	OrphaNet:808
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OrphaNet	OrphaNet:159
COL7A1	Generalized dominant dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:231568
COL7A1	Dystrophic epidermolysis bullosa pruriginosa	OrphaNet	OrphaNet:89843
COL7A1	Recessive dystrophic epidermolysis bullosa inversa	OrphaNet	OrphaNet:79409
COL7A1	Dystrophic epidermolysis bullosa, nails only	OrphaNet	OrphaNet:158676
COL7A1	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN	OrphaNet	OrphaNet:79411
COL7A1	Centripetalis recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:89841
COL7A1	Acral dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:158673
COL7A1	Recessive dystrophic epidermolysis bullosa-generalized other	OrphaNet	OrphaNet:89842

COL7A1	Epidermolysis bullosa simplex superficialis	OrphaNet	OrphaNet:89839	orphanet
COL7A1	Severe generalized recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79408	orphanet
COL7A1	Pretibial dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79410	orphanet
NDUFAF3	Isolated NADH-CoQ reductase deficiency	OrphaNet	OrphaNet:2609	orphanet
LAMB2	Synaptic congenital myasthenic syndromes	OrphaNet	OrphaNet:98915	orphanet
LAMB2	LAMB-2-related infantile-onset nephrotic syndrome	OrphaNet	OrphaNet:306507	orphanet
LAMB2	PIERSON SYNDROME	OrphaNet	OrphaNet:2670	orphanet
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899	OMIM
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	DECIPHER	MIM:212138	OMIM
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	DECIPHER	MIM:615760	OMIM

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)
IP6K2	missense variant	ENST00000449610	?	ENSP00000393077	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000431721	NM_001190316.1	ENSP00000414139	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000453202	?	ENSP00000387394	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000443964	?	ENSP00000410950	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000417896	?	ENSP00000388116	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000433104	?	ENSP00000396904	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000446860	NM_001190317.1	ENSP00000399052	G/D	gGt/gAt	?	?	1
QRICH1	missense variant	ENST00000424300	?	ENSP00000412890	S/N	aGt/aAt	?	?	1
QRICH1	missense variant	ENST00000395443	NM_198880.1	ENSP00000378830	S/N	aGt/aAt	?	?	1
QRICH1	missense variant	ENST00000357496	NM_017730.2	ENSP00000350094	S/N	aGt/aAt	?	?	1
USP19	missense variant	ENST00000453664	NM_001199162.1	ENSP00000400090	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000434032	NM_001199160.1	ENSP00000401197	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000398898	?	ENSP00000381872	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000398888	NM_006677.2	ENSP00000381863	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000398892	?	ENSP00000381867	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000417901	NM_001199161.1	ENSP00000395260	H/D	Cat/Gat	?	?	1
USP19	missense variant	ENST00000306026	?	ENSP00000303503	H/D	Cat/Gat	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
WDR6	?	ENSG00000178252	visceral adipocytes	1.53×10-7 (p-value)	GTEx Portal V6	105
NCKIPSD	?	ENSG00000213672	visceral adipocytes	2.83×10-7 (p-value)	GTEx Portal V6	69
WDR6	?	ENSG00000178252	stomach	2.40×10-7 (p-value)	GTEx Portal V6	103
CCDC36	?	ENSG00000173421	muscularis mucosae	1.49×10-7 (p-value)	GTEx Portal V6	92

KLHDC8B	?	ENSG00000185909	muscularis mucosae	8.00×10-6 (p-value)	GTEx Portal V6	10
WDR6	?	ENSG00000178252	lung	1.44×10-10 (p-value)	GTEx Portal V6	107
SLC26A6	?	ENSG00000225697	lung	9.39×10-8 (p-value)	GTEx Portal V6	105
CELSR3	?	ENSG00000008300	lung	2.18×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	cortex	1.66×10-6 (p-value)	GTEx Portal V6	69
WDR6	?	ENSG00000178252	tibial nerve	7.26×10-13 (p-value)	GTEx Portal V6	107
SEMA3F	?	ENSG00000001617	tibial nerve	1.42×10-6 (p-value)	GTEx Portal V6	24
WDR6	?	ENSG00000178252	transformed fibroblasts	7.13×10-18 (p-value)	GTEx Portal V6	107
CCDC36	?	ENSG00000173421	transformed fibroblasts	2.32×10-7 (p-value)	GTEx Portal V6	95
NCKIPSD	?	ENSG00000213672	transformed fibroblasts	3.12×10-8 (p-value)	GTEx Portal V6	95
IP6K2	?	ENSG00000068745	transformed fibroblasts	1.54×10-7 (p-value)	GTEx Portal V6	27
RP11-3B7.1	?	ENSG00000225399	transformed fibroblasts	4.31×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	tibial artery	3.97×10-9 (p-value)	GTEx Portal V6	107
TMEM89	?	ENSG00000183396	tibial artery	1.39×10-7 (p-value)	GTEx Portal V6	105
NICN1	?	ENSG00000145029	tibial artery	5.92×10-8 (p-value)	GTEx Portal V6	78
IP6K2	?	ENSG00000068745	tibial artery	6.46×10-7 (p-value)	GTEx Portal V6	27
AMT	?	ENSG00000145020	tibial artery	6.08×10-7 (p-value)	GTEx Portal V6	78
CCDC36	?	ENSG00000173421	tibial artery	4.02×10-5 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	blood	8.00×10-12 (p-value)	GTEx Portal V6	107
WDR6	?	ENSG00000178252	esophagus mucosa	7.00×10-34 (p-value)	GTEx Portal V6	107
NCKIPSD	?	ENSG00000213672	esophagus mucosa	8.96×10-9 (p-value)	GTEx Portal V6	107
CCDC36	?	ENSG00000173421	testis	8.22×10-8 (p-value)	GTEx Portal V6	106
WDR6	?	ENSG00000178252	skeletal muscle	2.27×10-13 (p-value)	GTEx Portal V6	107
SLC26A6	?	ENSG00000225697	skeletal muscle	3.53×10-10 (p-value)	GTEx Portal V6	73
NCKIPSD	?	ENSG00000213672	skeletal muscle	1.01×10-5 (p-value)	GTEx Portal V6	20
TMA7	?	ENSG00000232112	skeletal muscle	1.82×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	unexposed skin	1.27×10-26 (p-value)	GTEx Portal V6	107
WDR6	?	ENSG00000178252	sun exposed skin	7.41×10-50 (p-value)	GTEx Portal V6	107
RBM6	?	ENSG00000004534	sun exposed skin	1.41×10-5 (p-value)	GTEx Portal V6	38
WDR6	?	ENSG00000178252	subcutaneous adipocytes	2.01×10-21 (p-value)	GTEx Portal V6	107
ARIH2	?	ENSG00000177479	subcutaneous adipocytes	9.11×10-6 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	breast	2.94×10-7 (p-value)	GTEx Portal V6	75
WDR6	?	ENSG00000178252	EBV lymphocytes	9.15×10-7 (p-value)	GTEx Portal V6	27
SLC26A6	?	ENSG00000225697	EBV lymphocytes	6.39×10-6 (p-value)	GTEx Portal V6	27
IP6K2	?	ENSG00000068745	aorta	1.01×10-5 (p-value)	GTEx Portal V6	25
NICN1	?	ENSG00000145029	aorta	5.04×10-5 (p-value)	GTEx Portal V6	1

CCDC71	?	ENSG00000177352	cerebellar hemisphere	5.33×10^{-6} (p-value)	GTEx Portal V6	26
WDR6	ENST00000615452	ILMN_1669484	skin	7.40×10^{-45} (p-value)	MuTHER consortium	15
WDR6	ENST00000452875		adipocyte	8.90×10^{-42} (p-value)	MuTHER consortium	15
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	skin	1.46×10^{-7} (p-value)	MuTHER consortium	15
USP4	ENST00000483212		blood	7.52×10^{-5} (p-value)	MuTHER consortium	15
USP4	ENST00000485450		adipocyte	1.06×10^{-12} (p-value)	MuTHER consortium	15
C3orf62	ENST00000343010					
TMEM89	ENST00000330862	ILMN_1694339	skin	1.23×10^{-6} (p-value)	MuTHER consortium	14
RP11-3B7.1	ENST00000622888	ILMN_1807244	adipocyte	1.03×10^{-19} (p-value)	MuTHER consortium	15
RP11-3B7.1	ENST00000440528					
C3orf62	ENST00000343010	ILMN_1679339	blood	1.87×10^{-5} (p-value)	MuTHER consortium	7
C3orf62	ENST00000479673					
RBM6	ENST00000266022	ILMN_1738239	adipocyte	2.62×10^{-5} (p-value)	MuTHER consortium	2
RBM6	ENST00000442092					
RBM6	ENST00000454079					
RBM6	ENST00000419610					
RBM6	ENST00000422955					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
WDR6	ENST00000615452	ILMN_1669484	monocyte	1.45×10^{-16} (p-value)	Fairfax et al.	16
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
RBM6	ENST00000442092	ILMN_1738239	monocyte	8.89×10^{-5} (p-value)	Fairfax et al.	2
RBM6	ENST00000266022		b-cell	5.55×10^{-5} (p-value)	Fairfax et al.	2
RBM6	ENST00000454079					
RBM6	ENST00000422955					
RBM6	ENST00000419610					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
UBA7	ENST00000497908	ILMN_1794612	b-cell	6.47×10^{-5} (p-value)	Fairfax et al.	16
UBA7	ENST00000333486					
MIR5193	ENST00000584510					
USP4	ENST00000351842	ILMN_1798712	monocyte	1.23×10^{-11} (p-value)	Fairfax et al.	16
USP4	ENST00000483212		b-cell	1.99×10^{-8} (p-value)	Fairfax et al.	16
C3orf62	ENST00000343010					
USP4	ENST00000485450					
SLC26A6	ENST00000420764	ILMN_1785252	b-cell	3.59×10^{-5} (p-value)	Fairfax et al.	16
SLC26A6	ENST00000395550					
SLC26A6	ENST00000489483					

SLC26A6	ENST00000480524					
SLC26A6	ENST00000455886					
SLC26A6	ENST00000307364					
SLC26A6	ENST00000614797					
SLC26A6	ENST00000358747					
SLC26A6	ENST00000383733					
SLC26A6	ENST00000337000					
CCDC71	ENST00000321895	ILMN_1768433	monocyte	3.61×10^{-4} (p-value)	Fairfax et al.	1
			b-cell	2.91×10^{-4} (p-value)	Fairfax et al.	1
WDR6	?	ENSG00000178252	pancreas	3.37×10^{-5} (p-value)	GTEx Portal V6	1
WDR6	ENST00000615452	ILMN_1669484	monocyte	1.21×10^{-38} (p-value)	Zeller et al.	4
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	monocyte	6.52×10^{-34} (p-value)	Zeller et al.	4
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
WDR6	?	ENSG00000178252	terminal ileum	4.57×10^{-6} (p-value)	GTEx Portal V6	6
ARIH2	ENST00000356401	ILMN_1792825	blood	5.24×10^{-5} (p-value)	Westra et al.	3
RBM6	?	ENSG00000004534	thyroid	3.30×10^{-5} (p-value)	GTEx Portal V6	19

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000609678	2	ENST00000332780, ENST00000459846	KLHDC8B

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000036427		
ENCP00000036447		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000338847	1	ENCP00000036433	ARIH2
ENCE00000338585	1	ENCP00000036360	SPINK8
		ENCP00000036439	ARIH2
		ENCP00000036380	ATRIP
		ENCP00000036426	IP6K2
ENCE00000338754	1	ENCP00000036427	PRKAR2A
		ENCP00000036409	SLC26A6
ENCE00000338989	1	ENCP00000036489	CCDC36
ENCE00000338724	1	ENCP00000036395	UCN2

ENCE00000338727	1	ENCP00000036460	IMPDH2
		ENCP00000036397	COL7A1 COL7A1
		ENCP00000036451	DALRD3
ENCE00000338767	2	ENCP00000036414	CELSR3
		ENCP00000036418	NCKIPSD
		ENCP00000036450	WDR6
		ENCP00000036472	QARS
		ENCP00000036507	AMT AMT
		ENCP00000036475	USP19
ENCE00000338922	1	ENCP00000036453	DALRD3
ENCE00000338903	1	ENCP00000036445	P4HTM

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001478981 (promoter flanking region)	3	embryonic stem cell (H1ESC)	PolII, H3K9ac, H3K27ac, H3K4me2, H3K36me3, DNase1, H3K27me3
		HSMMtube	H3K79me2, H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H3K79me2, H3K27ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3, H3K27ac, DNase1
		blood (GM12878)	PolII, H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K27ac, H3K4me1
		NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K4me1, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001363512 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3, H3K79me2
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	H3K79me2, CTCF, H3K36me3
		blood (GM12878)	H3K36me3, H3K79me2
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001363515 (promoter flanking region)	1	NHLF	H3K4me1, H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
		muscle (HSMM)	H3K79me2, H3K36me3
		breast (HMEC)	H3K4me2, H3K4me1, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K36me3, H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K79me2, H3K36me3

		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H4K20me1, H3K79me2, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K4me1, DNase1, H3K36me3, H3K4me2, H3K27ac
ENSR00001363517 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3
		HSMMtube	DNase1, H3K79me2, H3K9ac, H3K4me2, H3K4me3, H3K36me3
		blood (K562)	HEY1, H2AZ, TAF1, ELF1, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, H3K79me2, H3K9ac, Yy1, H3K27ac, Max
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	DNase1, H3K36me3, H3K27me3, H3K4me3, H3K27ac, Gabp, Yy1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, DNase1, H3K79me2
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K27ac, H3K36me3, H3K9ac, DNase1, H3K4me3
		Osteobl	H3K27me3, H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	DNase1, H3K36me3, H3K27me3, PolII, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001363520 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H3K4me2
ENSR00001363530 (open chromatin region)	1	endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (K562)	DNase1
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00001478987 (open chromatin region)	1	muscle (HSMM)	DNase1
ENSR00001363541 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, H3K4me2, H3K4me3
		HSMMtube	H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K27ac, Max, H3K9ac, H3K79me2, H2AZ, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K36me3, DNase1
		liver (HepG2)	PolII, H3K79me2, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K4me2, H4K5ac, H3K36me3, H3K4me3
		blood (GM12878)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, PolII, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K36me3, H3K4me2, H3K4me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	PolII, DNase1, H3K79me2, H3K4me3, H3K27ac, Ini1, Nrf1, H3K9ac, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me2, PolII, DNase1, Cmyc, H3K4me3, Max
		A549	H3K4me3, H3K4me2, H3K9ac, H3K36me3
ENSR00001674797 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
ENSR00001674799 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
ENSR00001478995 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		blood (K562)	Egr1, H3K79me2, H3K36me3
		muscle (HSMM)	H3K79me2, H3K36me3
		liver (HepG2)	H3K79me2, H3K36me3
		blood (GM12878)	BATF, H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3, H3K4me1
		NHLF	H3K36me3

		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001363566 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K36me3, CTCF, PolII, Rad21, TAF7, H3K27ac, ATF3, Yy1, TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H2AZ, Max, Cfos, PolII, H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, Brg1, Egr1, H3K27ac, ZBTB33, H3K9ac, USF1, Cmyc, H3K79me2, HEY1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	DNase1, PolII, H3K79me2, TAF1, USF1, H3K4me1, ZBTB33, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K36me3, H3K27me3
		blood (GM12878)	PolII, H2AZ, DNase1, Egr1, CTCF, H3K79me2, H3K4me3, USF1, ZBTB33, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	H3K4me3, H3K9ac, CTCF, H3K36me3, H4K5ac, DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1, H3K27ac, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me2, CTCF, H3K9ac, DNase1, H3K27ac, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, H3K9ac, PolII, DNase1
		A549	H3K36me3, H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00001363576 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, Egr1, TAF7, Sin3Ak20, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3
		HSMMtube	CTCF, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, DNase1
		blood (K562)	ZBTB33, Max, Rad21, H3K9ac, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Yy1, Egr1, H3K27ac
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, CTCF, H3K4me3
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, Yy1, PolII, H3K79me2, Rad21, TAF1, ZBTB33, H3K4me2, H3K9ac, H3K27ac, CTCF, H3K4me3, H3K36me3
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, CTCF
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, Egr1, Yy1, H3K79me2, H3K4me3, ZBTB33, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K36me3, H3K27ac, DNase1, H3K4me3, CTCF, H3K9ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3, H3K4me2, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K36me3, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, PolII, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1
ENSR00001479006 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	DNase1
		blood (DND-41)	CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	PolII, CTCF
		liver (HepG2)	HNF4G, Rad21, HNF4A, CTCF
		blood (GM12878)	Tr4, PolII, Rad21, CTCF
		skin (NHEK)	CTCF
ENSR00001363588 (promoter)	2	embryonic stem cell (H1ESC)	H3K27ac, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H3K36me3, CTCF, H3K9ac, H3K4me2, H3K4me3, H2AZ

		blood (K562)	H3K27ac, Max, H3K4me1, Rad21, H3K9ac, Cmyc, H3K79me2, H2AZ, PolII, H3K4me2, CTCF, H3K36me3, DNase1, H3K4me3, Ini1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, H3K36me3, DNase1
		liver (HepG2)	PolII, Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, CTCF
		blood (GM12878)	DNase1, H2AZ, Yy1, Rad21, H3K4me3, H3K4me2, CTCF
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, DNase1
		skin (NHEK)	H3K36me3, CTCF, H3K4me3, H3K4me2, H3K9ac, DNase1
		NHLF	DNase1, H3K4me3, CTCF, H3K9ac, H3K36me3
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	CTCF
		breast (HMEC)	H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K4me3
		monocytes (Monocytes-CD14+)	CTCF, H3K4me2, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1, PolII, H3K4me2, Max, H3K4me3
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF
ENSR00001479009	1	NHLF	DNase1
(promoter flanking region)		HSMMtube	H3K27me3
		Osteobl	H3K36me3
		blood (K562)	DNase1, Nfe2
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1, Jund, Max
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	FOSL2, Jund, DNase1, H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	DNase1, H3K36me3
		skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC141002.1	upstream gene variant, downstream gene variant	1996	ENST00000619404	?	?	7
ARIH2	upstream gene variant, downstream gene variant	1458	ENST00000492077	?	?	4
ARIH2	upstream gene variant	2504	ENST00000488963	?	?	3
ARIH2	upstream gene variant, downstream gene variant	2520	ENST00000433170	?	ENSP00000406063	4
ARIH2	upstream gene variant	2477	ENST00000478224	?	?	3
ARIH2	upstream gene variant	2935	ENST00000487891	?	?	1
ARIH2	upstream gene variant	2479	ENST00000498314	?	?	3
ARIH2	downstream gene variant	4664	ENST00000469038	?	?	1
ARIH2	upstream gene variant	2512	ENST00000486316	?	?	3
ARIH2	downstream gene variant	2338	ENST00000470296	?	?	1
ARIH2	upstream gene variant	2492	ENST00000449376	?	ENSP00000403222	3
ARIH2	upstream gene variant	3230	ENST00000465217	?	?	1
ARIH2	upstream gene variant, downstream gene variant	162	ENST00000490095	?	?	4
ARIH2	upstream gene variant	942	ENST00000459976	?	?	1
ARIH2	upstream gene variant, downstream gene variant	1593	ENST00000463738	?	?	4
ARIH2	upstream gene variant	4358	ENST00000482342	?	?	1
ARIH2	upstream gene variant	2500	ENST00000482427	?	?	3

ARIH2	upstream gene variant	2986	ENST00000472640	?	?	2
ARIH2	upstream gene variant, downstream gene variant	2297	ENST00000452882	?	ENSP00000395560	4
ARIH2	downstream gene variant	3647	ENST00000466850	?	?	1
ARIH2	upstream gene variant	2485	ENST00000474618	?	?	3
ARIH2	upstream gene variant	423	ENST00000463204	?	?	1
ARIH2	upstream gene variant, downstream gene variant	2513	ENST00000449729	?	ENSP00000404838	4
ARIH2	upstream gene variant	43	ENST00000452385	?	ENSP00000403845	2
ARIH2	upstream gene variant	2551	ENST00000474936	?	?	3
ARIH2	upstream gene variant	1104	ENST00000495507	?	?	2
ARIH2	upstream gene variant	2477	ENST00000356401	NM_006321.2	ENSP00000348769	3
ARIH2	upstream gene variant, downstream gene variant	2297	ENST00000420814	?	ENSP00000397225	4
ARIH2	upstream gene variant, downstream gene variant	2498	ENST00000495761	?	?	4
ARIH2	upstream gene variant	2477	ENST00000430423	?	ENSP00000399788	3
ARIH2	upstream gene variant	2500	ENST00000483333	?	?	3
ARIH2OS	downstream gene variant, upstream gene variant	1444	ENST00000408959	NM_001123040.1	ENSP00000386193	4
C3orf84	downstream gene variant, upstream gene variant	1186	ENST00000443990	?	?	4
C3orf84	downstream gene variant, upstream gene variant	202	ENST00000432035	?	ENSP00000455944	4
C3orf84	downstream gene variant, upstream gene variant	166	ENST00000545770	NM_001080528.2	ENSP00000454903	4
CCDC36	upstream gene variant	3314	ENST00000438782	?	ENSP00000391788	1
CCDC36	upstream gene variant	3927	ENST00000452691	NM_001135197.1	ENSP00000407837	1
CCDC36	upstream gene variant	3937	ENST00000366429	?	ENSP00000403700	1
CCDC36	upstream gene variant	4044	ENST00000493870	?	?	1
CCDC36	upstream gene variant	2855	ENST00000296449	NM_178173.3	ENSP00000296449	1
CCDC71	downstream gene variant, upstream gene variant	3339	ENST00000321895	NM_022903.3	ENSP00000319006	3
DALRD3	upstream gene variant	4386	ENST00000492585	?	?	1
DALRD3	upstream gene variant	4384	ENST00000496568	?	ENSP00000485305	1
IMPDH2	upstream gene variant	1093	ENST00000442157	?	ENSP00000403502	2
IMPDH2	upstream gene variant	1998	ENST00000429182	?	ENSP00000393525	1
IMPDH2	upstream gene variant	2329	ENST00000462980	?	?	1
IMPDH2	upstream gene variant	2344	ENST00000491610	?	?	1
IMPDH2	upstream gene variant	1701	ENST00000472328	?	?	1
IMPDH2	downstream gene variant, upstream gene variant	1063	ENST00000496837	?	?	3
IMPDH2	upstream gene variant	1434	ENST00000484872	?	?	1
IMPDH2	downstream gene variant, upstream gene variant	1034	ENST00000485500	?	?	3
IMPDH2	upstream gene variant	1839	ENST00000463903	?	?	1
IMPDH2	upstream gene variant	1262	ENST00000481274	?	?	1
IMPDH2	upstream gene variant	1081	ENST00000326739	NM_000884.2	ENSP00000321584	2
IMPDH2	upstream gene variant	1484	ENST00000466147	?	?	1

IMPDH2	upstream gene variant	1484	ENST00000466147	?	?	1
IP6K2	downstream gene variant, upstream gene variant	195	ENST00000412795	?	ENSP00000389755	4
IP6K2	downstream gene variant	1429	ENST00000433104	?	ENSP00000396904	1
IP6K2	downstream gene variant, upstream gene variant	229	ENST00000412850	?	ENSP00000395819	4
IP6K2	downstream gene variant, upstream gene variant	228	ENST00000431721	NM_001190316.1	ENSP00000414139	4
IP6K2	downstream gene variant, upstream gene variant	83	ENST00000434860	?	ENSP00000388420	6
IP6K2	downstream gene variant, upstream gene variant	251	ENST00000476645	?	?	8
IP6K2	downstream gene variant, upstream gene variant	241	ENST00000416707	?	ENSP00000387759	4
IP6K2	downstream gene variant, upstream gene variant	81	ENST00000440424	?	ENSP00000393797	6
IP6K2	downstream gene variant, upstream gene variant	122	ENST00000424035	?	ENSP00000406357	6
IP6K2	downstream gene variant, upstream gene variant	238	ENST00000417896	?	ENSP00000388116	4
IP6K2	downstream gene variant, upstream gene variant	256	ENST00000454335	?	ENSP00000412121	4
IP6K2	downstream gene variant, upstream gene variant	1915	ENST00000455545	?	ENSP00000410454	4
IP6K2	downstream gene variant, upstream gene variant	166	ENST00000432678	NM_001146178.2, NM_001146179.2	ENSP00000400812	4
IP6K2	downstream gene variant, upstream gene variant	234	ENST00000413298	?	ENSP00000396203	4
IP6K2	downstream gene variant, upstream gene variant	278	ENST00000413654	?	ENSP00000414428	4
IP6K2	downstream gene variant, upstream gene variant	252	ENST00000443964	?	ENSP00000410950	4
IP6K2	downstream gene variant, upstream gene variant	82	ENST00000453202	?	ENSP00000387394	5
IP6K2	downstream gene variant, upstream gene variant	256	ENST00000436134	?	?	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000449563	?	ENSP00000411776	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000446860	NM_001190317.1	ENSP00000399052	4
IP6K2	downstream gene variant, upstream gene variant	252	ENST00000449610	?	ENSP00000393077	4
IP6K2	downstream gene variant, upstream gene variant	194	ENST00000328631	NM_001005909.2, NM_016291.3	ENSP00000331103	4
IP6K2	downstream gene variant, upstream gene variant	1957	ENST00000450045	?	ENSP00000394488	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000443853	?	ENSP00000389761	4
IP6K2	downstream gene variant, upstream gene variant	1455	ENST00000491686	?	?	5
IP6K2	downstream gene variant, upstream gene variant	228	ENST00000340879	NM_001005910.2, NM_001005911.2	ENSP00000341925	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000437427	?	ENSP00000403968	4
IP6K2	downstream gene variant, upstream gene variant	486	ENST00000479914	?	?	4
KLHDC8B	upstream gene variant, downstream gene variant	1195	ENST00000332780	NM_173546.2	ENSP00000327468	4
KLHDC8B	upstream gene variant, downstream gene variant	631	ENST00000476495	?	?	7
KLHDC8B	upstream gene variant, downstream gene variant	411	ENST00000471811	?	?	8
KLHDC8B	upstream gene variant, downstream gene variant	430	ENST00000459846	?	?	5
KLHDC8B	upstream gene variant, downstream gene variant	1533	ENST00000462582	?	?	6
LAMB2	downstream gene variant	4680	ENST00000480640	?	?	1
LAMB2	downstream gene variant	2075	ENST00000484713	?	?	1
LAMB2	downstream gene variant	4919	ENST00000538659	?	?	1

LAMB2	downstream gene variant	2074	ENST00000467506	?	?	1
LAMB2	downstream gene variant	2217	ENST00000498377	?	?	1
LAMB2	upstream gene variant	3867	ENST00000494831	?	ENSP00000444751	1
LAMB2	downstream gene variant, upstream gene variant	2093	ENST00000305544	?	ENSP00000307156	2
LAMB2	downstream gene variant	4285	ENST00000477225	?	?	1
LAMB2	downstream gene variant, upstream gene variant	2074	ENST00000418109	NM_002292.3	ENSP00000388325	2
LAMB2	downstream gene variant	3454	ENST00000469665	?	?	1
MIR6890	downstream gene variant, upstream gene variant	2059	ENST00000620931	?	?	4
NCKIPSD	upstream gene variant	1697	ENST00000416649	NM_184231.2	ENSP00000389059	1
NCKIPSD	upstream gene variant	1220	ENST00000453349	?	ENSP00000408588	1
NCKIPSD	upstream gene variant	1654	ENST00000454134	?	ENSP00000416144	1
NCKIPSD	upstream gene variant	1746	ENST00000439518	?	ENSP00000409675	1
NCKIPSD	upstream gene variant	1654	ENST00000294129	NM_016453.3	ENSP00000294129	1
NCKIPSD	upstream gene variant	1217	ENST00000426678	?	ENSP00000416904	1
NDUFAF3	downstream gene variant	3390	ENST00000326912	NM_199074.1	ENSP00000323003	1
NDUFAF3	downstream gene variant	3182	ENST00000326925	NM_199069.1	ENSP00000323076	1
NDUFAF3	downstream gene variant	3501	ENST00000480392	?	?	1
NDUFAF3	downstream gene variant	3390	ENST00000496152	?	?	1
NDUFAF3	downstream gene variant	3390	ENST00000395458	NM_199073.1	ENSP00000378843	1
NDUFAF3	downstream gene variant	3389	ENST00000451378	NM_199070.1	ENSP00000402465	1
P4HTM	downstream gene variant	2685	ENST00000472796	?	ENSP00000476858	1
P4HTM	downstream gene variant	165	ENST00000491739	?	ENSP00000476303	1
P4HTM	downstream gene variant	165	ENST00000484115	?	?	1
P4HTM	downstream gene variant	2719	ENST00000609406	?	?	1
P4HTM	downstream gene variant	165	ENST00000343546	NM_177938.2	ENSP00000341422	1
P4HTM	downstream gene variant	4304	ENST00000485210	?	?	1
P4HTM	downstream gene variant	126	ENST00000383729	NM_177939.2	ENSP00000373235	1
P4HTM	downstream gene variant	3017	ENST00000444213	?	ENSP00000398554	1
P4HTM	upstream gene variant, downstream gene variant	1180	ENST00000486817	?	?	2
P4HTM	downstream gene variant	3085	ENST00000468374	?	?	1
P4HTM	downstream gene variant	165	ENST00000472301	?	?	1
PRKAR2A	upstream gene variant	700	ENST00000437821	?	ENSP00000400840	2
PRKAR2A	upstream gene variant	1748	ENST00000265563	NM_004157.2	ENSP00000265563	1
PRKAR2A	upstream gene variant	1790	ENST00000454963	?	ENSP00000394041	1
PRKAR2A	upstream gene variant	1761	ENST00000419216	?	ENSP00000411432	1
PRKAR2A	upstream gene variant	1766	ENST00000296446	?	ENSP00000296446	1
PRKAR2A-AS1	upstream gene variant, downstream gene variant	158	ENST00000431705	?	?	2

PRKAR2A-AS1	downstream gene variant	1101	ENST00000435419	?	?	1
PRKAR2A-AS1	upstream gene variant, downstream gene variant	140	ENST00000412171	?	?	2
PRKAR2A-AS1	upstream gene variant, downstream gene variant	178	ENST00000416209	?	?	2
QARS	downstream gene variant, upstream gene variant	940	ENST00000470225	?	?	2
QARS	downstream gene variant	730	ENST00000417025	?	?	1
QARS	downstream gene variant	693	ENST00000418549	?	ENSP00000415247	1
QARS	downstream gene variant, upstream gene variant	1306	ENST00000478561	?	?	4
QARS	downstream gene variant	4624	ENST00000452739	?	ENSP00000392850	1
QARS	downstream gene variant	332	ENST00000482438	?	?	2
QARS	downstream gene variant	634	ENST00000479495	?	?	1
QARS	downstream gene variant	4832	ENST00000482261	?	?	1
QARS	downstream gene variant, upstream gene variant	73	ENST00000494767	?	?	3
QARS	downstream gene variant	226	ENST00000470619	?	?	1
QARS	downstream gene variant, upstream gene variant	1552	ENST00000482468	?	?	4
QARS	downstream gene variant, upstream gene variant	827	ENST00000459870	?	?	3
QARS	downstream gene variant	818	ENST00000464962	?	?	1
QARS	downstream gene variant	812	ENST00000430182	?	ENSP00000389823	1
QARS	downstream gene variant, upstream gene variant	813	ENST00000494984	?	?	3
QARS	downstream gene variant	3193	ENST00000494838	?	?	1
QARS	downstream gene variant, upstream gene variant	873	ENST00000466179	?	?	3
QARS	downstream gene variant, upstream gene variant	812	ENST00000487495	?	?	2
QARS	downstream gene variant, upstream gene variant	811	ENST00000453392	?	ENSP00000396326	3
QARS	downstream gene variant, upstream gene variant	812	ENST00000475599	?	?	2
QARS	downstream gene variant, upstream gene variant	811	ENST00000482248	?	?	3
QARS	downstream gene variant	816	ENST00000414533	NM_001272073.1	ENSP00000390015	1
QARS	downstream gene variant, upstream gene variant	812	ENST00000497635	?	?	2
QARS	downstream gene variant, upstream gene variant	840	ENST00000470113	?	?	3
QARS	downstream gene variant	811	ENST00000306125	NM_005051.2	ENSP00000307567	1
QRICH1	downstream gene variant	2612	ENST00000479449	?	?	1
QRICH1	downstream gene variant, upstream gene variant	411	ENST00000437939	?	ENSP00000416133	6
QRICH1	downstream gene variant, upstream gene variant	849	ENST00000469910	?	?	3
QRICH1	downstream gene variant, upstream gene variant	411	ENST00000450685	?	ENSP00000413051	6
QRICH1	downstream gene variant, upstream gene variant	411	ENST00000411682	?	ENSP00000412870	6
QRICH1	downstream gene variant, upstream gene variant	577	ENST00000430979	?	ENSP00000405505	6
QRICH1	downstream gene variant, upstream gene variant	816	ENST00000477021	?	?	3
QRICH1	downstream gene variant, upstream gene variant	758	ENST00000395443	NM_198880.1	ENSP00000378830	3
QRICH1	downstream gene variant, upstream gene variant	2576	ENST00000498440	?	?	3

QRICH1	downstream gene variant, upstream gene variant	187	ENST00000498392	?	?	4
QRICH1	downstream gene variant, upstream gene variant	1059	ENST00000357496	NM_017730.2	ENSP00000350094	3
QRICH1	downstream gene variant	3698	ENST00000489642	?	?	1
QRICH1	downstream gene variant, upstream gene variant	1510	ENST00000424300	?	ENSP00000412890	3
RN7SL182P	upstream gene variant, downstream gene variant	3852	ENST00000496499	?	?	2
RP11-694I15.7	downstream gene variant	3163	ENST00000603877	?	?	1
RP13-131K19.1	downstream gene variant	143	ENST00000415982	?	?	1
RP13-131K19.1	downstream gene variant	157	ENST00000429681	?	?	1
RP13-131K19.2	upstream gene variant	4960	ENST00000452042	?	?	1
RP13-131K19.6	upstream gene variant, downstream gene variant	765	ENST00000607245	?	?	3
SLC25A20	downstream gene variant	1228	ENST00000430379	?	ENSP00000388986	1
SLC25A20	downstream gene variant	1233	ENST00000319017	NM_000387.5	ENSP00000326305	1
SLC25A20	downstream gene variant	1689	ENST00000440964	?	ENSP00000388563	1
SLC25A20	downstream gene variant	1538	ENST00000479050	?	?	1
USP19	downstream gene variant, upstream gene variant	1062	ENST00000398896	?	ENSP00000381870	2
USP19	downstream gene variant	4367	ENST00000417901	NM_001199161.1	ENSP00000395260	1
USP19	downstream gene variant	4363	ENST00000398898	?	ENSP00000381872	1
USP19	downstream gene variant	4990	ENST00000398892	?	ENSP00000381867	1
USP19	downstream gene variant	4992	ENST00000398888	NM_006677.2	ENSP00000381863	1
USP19	upstream gene variant	1329	ENST00000464931	?	?	1
USP19	upstream gene variant	1252	ENST00000480163	?	?	1
USP19	downstream gene variant	4448	ENST00000453664	NM_001199162.1	ENSP00000400090	1
USP19	upstream gene variant	1682	ENST00000479073	?	ENSP00000428539	1
WDR6	upstream gene variant	4354	ENST00000615452	?	ENSP00000482289	1
WDR6	upstream gene variant	117	ENST00000452875	?	ENSP00000414157	1
WDR6	upstream gene variant	152	ENST00000627177	?	ENSP00000487056	1
WDR6	upstream gene variant	123	ENST00000471162	?	?	1
WDR6	upstream gene variant	113	ENST00000438660	?	ENSP00000387692	1
WDR6	upstream gene variant	331	ENST00000419837	?	ENSP00000390740	1
WDR6	upstream gene variant	328	ENST00000472878	?	?	1
WDR6	upstream gene variant	82	ENST00000488572	?	?	1
WDR6	upstream gene variant	73	ENST00000429900	?	ENSP00000477418	1
WDR6	upstream gene variant	113	ENST00000608424	?	ENSP00000477389	1
WDR6	upstream gene variant	345	ENST00000461687	?	?	1
WDR6	upstream gene variant	113	ENST00000473238	?	?	1
WDR6	upstream gene variant	123	ENST00000420783	?	ENSP00000395993	1
WDR6	upstream gene variant	110	ENST00000462064	?	?	1

WDR6	upstream gene variant	116	ENST00000415265 ?	ENSP00000412195	1
WDR6	upstream gene variant	328	ENST00000448293 ?	ENSP00000413432	1
WDR6	upstream gene variant	786	ENST00000491365 ?	ENSP00000477463	1
WDR6	upstream gene variant	781	ENST00000489427 ?	ENSP00000477087	1
WDR6	upstream gene variant	110	ENST00000489684 ?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
C3orf84	ENST00000545770	NM_001080528.2	ENSP00000454903	P	ccA/ccG	1
C3orf84	ENST00000432035	?	ENSP00000455944	P	ccA/ccG	1
QARS	ENST00000306125	NM_005051.2	ENSP00000307567	H	caT/caC	1
QARS	ENST00000414533	NM_001272073.1	ENSP00000390015	H	caT/caC	1
QARS	ENST00000452739	?	ENSP00000392850	H	caT/caC	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
KLHDC8B	ENST00000332780	NM_173546.2	ENSP00000327468	1
KLHDC8B	ENST00000462582	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ARIH2	ENST00000495507	?	?	4
ARIH2	ENST00000472640	?	?	1
ARIH2	ENST00000449729	?	ENSP00000404838	2
ARIH2	ENST00000495761	?	?	7
ARIH2	ENST00000452385	?	ENSP00000403845	6
ARIH2	ENST00000484999	?	?	7
ARIH2	ENST00000463204	?	?	5
ARIH2	ENST00000498314	?	?	7
ARIH2	ENST00000466850	?	?	2
ARIH2	ENST00000474936	?	?	7
ARIH2	ENST00000420814	?	ENSP00000397225	6
ARIH2	ENST00000463738	?	?	1
ARIH2	ENST00000433170	?	ENSP00000406063	2
ARIH2	ENST00000482427	?	?	7
ARIH2	ENST00000430423	?	ENSP00000399788	7
ARIH2	ENST00000470296	?	?	6
ARIH2	ENST00000452882	?	ENSP00000395560	6
ARIH2	ENST00000490095	?	?	7
ARIH2	ENST00000449376	?	ENSP00000403222	8
ARIH2	ENST00000478224	?	?	7

ARIH2	ENST00000474618	?	?	7
ARIH2	ENST00000356401	NM_006321.2	ENSP00000348769	8
ARIH2	ENST00000488963	?	?	7
ARIH2	ENST00000486316	?	?	7
ARIH2	ENST00000492077	?	?	1
ARIH2	ENST00000483333	?	?	7
C3orf84	ENST00000443990	?	?	2
C3orf84	ENST00000545770	NM_001080528.2	ENSP00000454903	3
C3orf84	ENST00000432035	?	ENSP00000455944	3
CCDC36	ENST00000452691	NM_001135197.1	ENSP00000407837	12
CCDC36	ENST00000296449	NM_178173.3	ENSP00000296449	12
CCDC36	ENST00000438782	?	ENSP00000391788	12
CCDC36	ENST00000366429	?	ENSP00000403700	12
CCDC36	ENST00000493870	?	?	12
CELSR3	ENST00000164024	NM_001407.2	ENSP00000164024	1
IMPDH2	ENST00000326739	NM_000884.2	ENSP00000321584	1
IMPDH2	ENST00000462980	?	?	1
IMPDH2	ENST00000429182	?	ENSP00000393525	1
IMPDH2	ENST00000442157	?	ENSP00000403502	1
IMPDH2	ENST00000491610	?	?	1
IP6K2	ENST00000431721	NM_001190316.1	ENSP00000414139	17
IP6K2	ENST00000440424	?	ENSP00000393797	16
IP6K2	ENST00000432678	NM_001146178.2, NM_001146179.2	ENSP00000400812	18
IP6K2	ENST00000412850	?	ENSP00000395819	18
IP6K2	ENST00000436134	?	?	18
IP6K2	ENST00000434860	?	ENSP00000388420	16
IP6K2	ENST00000446860	NM_001190317.1	ENSP00000399052	17
IP6K2	ENST00000437427	?	ENSP00000403968	18
IP6K2	ENST00000433104	?	ENSP00000396904	27
IP6K2	ENST00000413298	?	ENSP00000396203	18
IP6K2	ENST00000449610	?	ENSP00000393077	17
IP6K2	ENST00000424035	?	ENSP00000406357	16
IP6K2	ENST00000476645	?	?	13
IP6K2	ENST00000454335	?	ENSP00000412121	18
IP6K2	ENST00000450045	?	ENSP00000394488	18
IP6K2	ENST00000412795	?	ENSP00000389755	18
IP6K2	ENST00000443853	?	ENSP00000389761	18
IP6K2	ENST00000413654	?	ENSP00000414428	18
IP6K2	ENST00000340879	NM_001005910.2, NM_001005911.2	ENSP00000341925	18
IP6K2	ENST00000449563	?	ENSP00000411776	18
IP6K2	ENST00000417896	?	ENSP00000388116	17
IP6K2	ENST00000491686	?	?	1

IP6K2	ENST00000455545	?	ENSP00000410454	18
IP6K2	ENST00000328631	NM_001005909.2, NM_016291.3	ENSP00000331103	19
IP6K2	ENST00000443964	?	ENSP00000410950	17
IP6K2	ENST00000416707	?	ENSP00000387759	18
IP6K2	ENST00000479914	?	?	3
KLHDC8B	ENST00000332780	NM_173546.2	ENSP00000327468	1
KLHDC8B	ENST00000459846	?	?	1
KLHDC8B	ENST00000476495	?	?	1
P4HTM	ENST00000468374	?	?	1
P4HTM	ENST00000472301	?	?	1
P4HTM	ENST00000475629	?	ENSP00000477191	1
P4HTM	ENST00000343546	NM_177938.2	ENSP00000341422	1
P4HTM	ENST00000444213	?	ENSP00000398554	1
P4HTM	ENST00000484115	?	?	1
P4HTM	ENST00000383729	NM_177939.2	ENSP00000373235	1
PRKAR2A	ENST00000265563	NM_004157.2	ENSP00000265563	8
PRKAR2A	ENST00000419216	?	ENSP00000411432	6
PRKAR2A	ENST00000454963	?	ENSP00000394041	8
PRKAR2A	ENST00000437821	?	ENSP00000400840	2
PRKAR2A	ENST00000296446	?	ENSP00000296446	8
QARS	ENST00000430182	?	ENSP00000389823	2
QARS	ENST00000494838	?	?	1
QARS	ENST00000470113	?	?	1
QARS	ENST00000453392	?	ENSP00000396326	1
QARS	ENST00000497635	?	?	1
QARS	ENST00000466179	?	?	1
QARS	ENST00000306125	NM_005051.2	ENSP00000307567	2
QARS	ENST00000475599	?	?	1
QARS	ENST00000482248	?	?	1
QARS	ENST00000459870	?	?	1
QARS	ENST00000414533	NM_001272073.1	ENSP00000390015	2
QARS	ENST00000487495	?	?	1
QARS	ENST00000464962	?	?	2
QARS	ENST00000494984	?	?	1
QARS	ENST00000470225	?	?	1
QARS	ENST00000452739	?	ENSP00000392850	1
QARS	ENST00000482261	?	?	1
QRICH1	ENST00000450685	?	ENSP00000413051	2
QRICH1	ENST00000437939	?	ENSP00000416133	2
QRICH1	ENST00000430979	?	ENSP00000405505	2
QRICH1	ENST00000411682	?	ENSP00000412870	2

QRICH1	ENST00000498440	?	?	3
QRICH1	ENST00000424300	?	ENSP00000412890	11
QRICH1	ENST00000395443	NM_198880.1	ENSP00000378830	11
QRICH1	ENST00000479449	?	?	5
QRICH1	ENST00000357496	NM_017730.2	ENSP00000350094	11
QRICH1	ENST00000489642	?	?	3
RP11-694I15.7	ENST00000603877	?	?	3
SLC25A20	ENST00000440964	?	ENSP00000388563	3
SLC25A20	ENST00000319017	NM_000387.5	ENSP00000326305	3
SLC25A20	ENST00000430379	?	ENSP00000388986	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IP6K2	ENST00000431721	NM_001190316.1	ENSP00000414139	1
IP6K2	ENST00000446860	NM_001190317.1	ENSP00000399052	1
IP6K2	ENST00000433104	?	ENSP00000396904	1
IP6K2	ENST00000449610	?	ENSP00000393077	1
IP6K2	ENST00000412795	?	ENSP00000389755	1
IP6K2	ENST00000417896	?	ENSP00000388116	1
IP6K2	ENST00000443964	?	ENSP00000410950	1
IP6K2	ENST00000416707	?	ENSP00000387759	1
QARS	ENST00000430182	?	ENSP00000389823	1
QARS	ENST00000418549	?	ENSP00000415247	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IP6K2	ENST00000433104	?	ENSP00000396904	1
KLHDC8B	ENST00000332780	NM_173546.2	ENSP00000327468	2
PRKAR2A	ENST00000265563	NM_004157.2	ENSP00000265563	1
PRKAR2A	ENST00000419216	?	ENSP00000411432	1
PRKAR2A	ENST00000454963	?	ENSP00000394041	1
PRKAR2A	ENST00000296446	?	ENSP00000296446	1
WDR6	ENST00000610967	NM_018031.3	ENSP00000484799	1
WDR6	ENST00000395474	?	ENSP00000378857	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C3orf84	ENST00000443990	?	1
IP6K2	ENST00000436134	?	1
KLHDC8B	ENST00000459846	?	2
PRKAR2A-AS1	ENST00000435419	?	1
PRKAR2A-AS1	ENST00000416209	?	1
QARS	ENST00000494838	?	1
QARS	ENST00000417025	?	1

QARS	ENST00000464962	?	1
QARS	ENST00000470619	?	1
QARS	ENST00000479495	?	1
QARS	ENST00000482261	?	1
QRICH1	ENST00000498392	?	1
QRICH1	ENST00000489642	?	1
USP19	ENST00000465902	?	1
USP19	ENST00000488993	?	1
USP19	ENST00000491859	?	1