

SNiPACard

Block annotations

Block info

genomic range	chr3:48,688,114-49,269,270 <i>e!</i>
block size	581,157 bp
variant count	107 variants

Basic features

Conservation/deleteriousness		Linked genes
phyloP	$\mu = -0.089$ [-2.931 – 4.247]	AC141002.1 <i>e!</i> , ARIH2 <i>e!</i> , ARIH2OS <i>e!</i> , C3orf84 <i>e!</i> , CCDC36 <i>e!</i> , CCDC71 <i>e!</i> , CELSR3 <i>e!</i> , DALRD3 <i>e!</i> , IMPDH2 <i>e!</i> , IP6K2 <i>e!</i> , KLHDC8B <i>e!</i> , LAMB2 <i>e!</i> , MIR6890 <i>e!</i> , NCKIPSD <i>e!</i> , NDUFAF3 <i>e!</i> , P4HTM <i>e!</i> , PRKAR2A <i>e!</i> , PRKAR2A-AS1 <i>e!</i> , QARS <i>e!</i> , QRIC1 <i>e!</i> , RN7SL182P <i>e!</i> , RP11-694115.7 <i>e!</i> , RP13-131K19.1 <i>e!</i> , RP13-131K19.2 <i>e!</i> , RP13-131K19.6 <i>e!</i> , SLC25A20 <i>e!</i> , USP19 <i>e!</i> , WDR6 <i>e!</i>
phastCons	$\mu = 0.113$ [0 – 1]	AMT <i>e!</i> , ARIH2 <i>e!</i> , C3orf62 <i>e!</i> , CCDC36 <i>e!</i> , CCDC71 <i>e!</i> , CELSR3 <i>e!</i> , IP6K2 <i>e!</i> , KLHDC8B <i>e!</i> , MIR5193 <i>e!</i> , NCKIPSD <i>e!</i> , NICN1 <i>e!</i> , RBM6 <i>e!</i> , RP11-3B7.1 <i>e!</i> , SEMA3F <i>e!</i> , SLC26A6 <i>e!</i> , TMA7 <i>e!</i> , TMEM89 <i>e!</i> , UBA7 <i>e!</i> , USP4 <i>e!</i> , WDR6 <i>e!</i>
GERP++	$\mu = -0.294$ [-10 – 4.95]	AMT <i>e!</i> , AMT <i>e!</i> , ARIH2 <i>e!</i> , ATRIP <i>e!</i> , CCDC36 <i>e!</i> , CELSR3 <i>e!</i> , COL7A1 <i>e!</i> , COL7A1 <i>e!</i> , DALRD3 <i>e!</i> , IMPDH2 <i>e!</i> , IP6K2 <i>e!</i> , KLHDC8B <i>e!</i> , NCKIPSD <i>e!</i> , P4HTM <i>e!</i> , PRKAR2A <i>e!</i> , QARS <i>e!</i> , SLC26A6 <i>e!</i> , SPINK8 <i>e!</i> , UCN2 <i>e!</i> , USP19 <i>e!</i> , WDR6 <i>e!</i>
CADD score	$\mu = 5.047$ [0.048 – 21.7]	KLHDC8B <i>e!</i> , AMT <i>e!</i> , SLC25A20 <i>e!</i> , COL7A1 <i>e!</i> , NDUFAF3 <i>e!</i> , QARS <i>e!</i> , LAMB2 <i>e!</i> , ATRIP <i>e!</i>

Trait annotations

Disease gene annotation

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

COL7A1 <i>e!</i>	Epidermolysis bullosa simplex superficialis	OrphaNet	OrphaNet:89839	orphanet
COL7A1 <i>e!</i>	Severe generalized recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79408	orphanet
COL7A1 <i>e!</i>	Pretibial dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79410	orphanet
NDUFAF3 <i>e!</i>	Isolated NADH-CoQ reductase deficiency	OrphaNet	OrphaNet:2609	orphanet
LAMB2 <i>e!</i>	Synaptic congenital myasthenic syndromes	OrphaNet	OrphaNet:98915	orphanet
LAMB2 <i>e!</i>	LAMB-2-related infantile-onset nephrotic syndrome	OrphaNet	OrphaNet:306507	orphanet
LAMB2 <i>e!</i>	PIERSON SYNDROME	OrphaNet	OrphaNet:2670	orphanet
AMT <i>e!</i>	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899	OMIM®
SLC25A20 <i>e!</i>	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	DECIPHER	MIM:212138	OMIM®
QARS <i>e!</i>	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 <i>e!</i>	missense variant	ENST00000449610 ?		ENSP000000393077	G/D	gGt/gAt	?	?	1
IP6K2 <i>e!</i>	missense variant	ENST00000431721	NM_001190316.1	ENSP000000414139	G/D	gGt/gAt	?	?	1
IP6K2 <i>e!</i>	missense variant	ENST00000453202 ?		ENSP000000387394	G/D	gGt/gAt	?	?	1
IP6K2 <i>e!</i>	missense variant	ENST00000443964 ?		ENSP000000410950	G/D	gGt/gAt	?	?	1
IP6K2 <i>e!</i>	missense variant	ENST00000417896 ?		ENSP000000388116	G/D	gGt/gAt	?	?	1
IP6K2 <i>e!</i>	missense variant	ENST00000433104 ?		ENSP000000396904	G/D	gGt/gAt	?	?	1
IP6K2 <i>e!</i>	missense variant	ENST00000446860	NM_001190317.1	ENSP000000399052	G/D	gGt/gAt	?	?	1
QRICH1 <i>e!</i>	missense variant	ENST00000424300 ?		ENSP000000412890	S/N	aGt/aAt	?	?	1
QRICH1 <i>e!</i>	missense variant	ENST00000395443	NM_198880.1	ENSP000000378830	S/N	aGt/aAt	?	?	1
QRICH1 <i>e!</i>	missense variant	ENST00000357496	NM_017730.2	ENSP000000350094	S/N	aGt/aAt	?	?	1
USP19 <i>e!</i>	missense variant	ENST00000453664	NM_001199162.1	ENSP000000400090	H/D	Cat/Gat	?	?	1
USP19 <i>e!</i>	missense variant	ENST00000434032	NM_001199160.1	ENSP000000401197	H/D	Cat/Gat	?	?	1
USP19 <i>e!</i>	missense variant	ENST00000398898 ?		ENSP000000381872	H/D	Cat/Gat	?	?	1
USP19 <i>e!</i>	missense variant	ENST00000398888	NM_006677.2	ENSP000000381863	H/D	Cat/Gat	?	?	1
USP19 <i>e!</i>	missense variant	ENST00000398892 ?		ENSP000000381867	H/D	Cat/Gat	?	?	1
USP19 <i>e!</i>	missense variant	ENST00000417901	NM_001199161.1	ENSP000000395260	H/D	Cat/Gat	?	?	1
USP19 <i>e!</i>	missense variant	ENST00000306026 ?		ENSP000000303503	H/D	Cat/Gat	?	?	1

Direct effect on regulation

cis-eQTL

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

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

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

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

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000609678 <i>e!</i>	2	ENST00000332780 <i>e!</i> , ENST00000459846 <i>e!</i>	KLHDC8B <i>e!</i>

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000036427 <i>e!</i>		
ENCP00000036447 <i>e!</i>		

ENCODE promoter-associated distal DHS (Enhancer)

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

ENCE00000338727 <i>e!</i>	1	ENCP00000036460 ENCP00000036397	IMPDH2 <i>e!</i> COL7A1 <i>e!</i> COL7A1 <i>e!</i>
ENCE00000338767 <i>e!</i>	2	ENCP00000036451 ENCP00000036414 ENCP00000036418 ENCP00000036450 ENCP00000036472 ENCP00000036507	DALRD3 <i>e!</i> CELSR3 <i>e!</i> NCKIPSD <i>e!</i> WDR6 <i>e!</i> QARS <i>e!</i> AMT <i>e!</i> AMT <i>e!</i>
ENCE00000338922 <i>e!</i>	1	ENCP00000036453	USP19 <i>e!</i> DALRD3 <i>e!</i>
ENCE00000338903 <i>e!</i>	1	ENCP00000036445	P4HTM <i>e!</i>

Regulatory feature cluster

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

		blood (GM12878)	H3K79me2, H3K36me3	
		lung (IMR90)	H4K20me1, H3K79me2, H3K36me3	
		A549	H3K36me3	
		skin (NHEK)	H3K4me1, DNase1, H3K36me3, H3K4me2, H3K27ac	
ENSR00001363517	e!	2	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3
(promoter flanking region)			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	e!	1	embryonic stem cell (H1ESC)	H3K36me3
(enhancer)			liver (HepG2)	H3K4me2
ENSR00001363530	e!	1	endothelium (HUVEC)	H3K36me3
(open chromatin region)			lung (IMR90)	H3K36me3
			blood (K562)	DNase1
			skin (NHEK)	H3K36me3
			muscle (HSMM)	H3K36me3
ENSR00001478987	e!	1	muscle (HSMM)	DNase1
(open chromatin region)				
ENSR00001363541	e!	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, H3K4me2, H3K4me3
(promoter)			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	e!	1	embryonic stem cell (H1ESC)	DNase1
(open chromatin region)				
ENSR00001674799	e!	1	monocytes (Monocytes-CD14+)	H3K36me3
(enhancer)				
ENSR00001478995	e!	1	embryonic stem cell (H1ESC)	H3K36me3
(enhancer)			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 <i>e!</i>	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K36me3, CTCF, PolII, Rad21, TAF7, H3K27ac, ATF3, Yy1, TAF1, H3K4me2, SP1, USF1, H3K9ac, H3K4me3
(promoter)		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 <i>e!</i>	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, Egr1, TAF7, Sin3Ak20, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3
(promoter)		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 <i>e!</i>	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
(CTCF binding site)		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 <i>e!</i>	2	embryonic stem cell (H1ESC)	H3K27ac, Rad21, CTCF, H3K4me2, H3K4me3, DNase1
(promoter)		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 <i>e!</i> 1 (promoter flanking region)	NHLF	DNase1
	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 <i>e!</i>	upstream gene variant, downstream gene variant	1996	ENST00000619404 ? <i>e!</i>		?	7
ARIH2 <i>e!</i>	upstream gene variant, downstream gene variant	1458	ENST00000492077 ? <i>e!</i>		?	4
ARIH2 <i>e!</i>	upstream gene variant	2504	ENST00000488963 ? <i>e!</i>		?	3
ARIH2 <i>e!</i>	upstream gene variant, downstream gene variant	2520	ENST00000433170 ? <i>e!</i>		ENSP00000406063 <i>e!</i>	4
ARIH2 <i>e!</i>	upstream gene variant	2477	ENST00000478224 ? <i>e!</i>		?	3
ARIH2 <i>e!</i>	upstream gene variant	2935	ENST00000487891 ? <i>e!</i>		?	1
ARIH2 <i>e!</i>	upstream gene variant	2479	ENST00000498314 ? <i>e!</i>		?	3
ARIH2 <i>e!</i>	downstream gene variant	4664	ENST00000469038 ? <i>e!</i>		?	1
ARIH2 <i>e!</i>	upstream gene variant	2512	ENST00000486316 ? <i>e!</i>		?	3
ARIH2 <i>e!</i>	downstream gene variant	2338	ENST00000470296 ? <i>e!</i>		?	1
ARIH2 <i>e!</i>	upstream gene variant	2492	ENST00000449376 ? <i>e!</i>		ENSP00000403222 <i>e!</i>	3
ARIH2 <i>e!</i>	upstream gene variant	3230	ENST00000465217 ? <i>e!</i>		?	1
ARIH2 <i>e!</i>	upstream gene variant, downstream gene variant	162	ENST00000490095 ? <i>e!</i>		?	4
ARIH2 <i>e!</i>	upstream gene variant	942	ENST00000459976 ? <i>e!</i>		?	1
ARIH2 <i>e!</i>	upstream gene variant, downstream gene variant	1593	ENST00000463738 ? <i>e!</i>		?	4
ARIH2 <i>e!</i>	upstream gene variant	4358	ENST00000482342 ? <i>e!</i>		?	1
ARIH2 <i>e!</i>	upstream gene variant	2500	ENST00000482427 ? <i>e!</i>		?	3

ARIH2 <i>e!</i>	upstream gene variant	2986	ENST00000472640 ?	?	2
ARIH2 <i>e!</i>	upstream gene variant, downstream gene variant	2297	ENST00000452882 ?	ENSP00000395560	4
ARIH2 <i>e!</i>	downstream gene variant	3647	ENST00000466850 ?	?	1
ARIH2 <i>e!</i>	upstream gene variant	2485	ENST00000474618 ?	?	3
ARIH2 <i>e!</i>	upstream gene variant	423	ENST00000463204 ?	?	1
ARIH2 <i>e!</i>	upstream gene variant, downstream gene variant	2513	ENST00000449729 ?	ENSP00000404838	4
ARIH2 <i>e!</i>	upstream gene variant	43	ENST00000452385 ?	ENSP00000403845	2
ARIH2 <i>e!</i>	upstream gene variant	2551	ENST00000474936 ?	?	3
ARIH2 <i>e!</i>	upstream gene variant	1104	ENST00000495507 ?	?	2
ARIH2 <i>e!</i>	upstream gene variant	2477	ENST00000356401 NM_006321.2	ENSP00000348769	3
ARIH2 <i>e!</i>	upstream gene variant, downstream gene variant	2297	ENST00000420814 ?	ENSP00000397225	4
ARIH2 <i>e!</i>	upstream gene variant, downstream gene variant	2498	ENST00000495761 ?	?	4
ARIH2 <i>e!</i>	upstream gene variant	2477	ENST00000430423 ?	ENSP00000399788	3
ARIH2 <i>e!</i>	upstream gene variant	2500	ENST00000483333 ?	?	3
ARIH2OS <i>e!</i>	downstream gene variant, upstream gene variant	1444	ENST00000408959 NM_001123040.1	ENSP00000386193	4
C3orf84 <i>e!</i>	downstream gene variant, upstream gene variant	1186	ENST00000443990 ?	?	4
C3orf84 <i>e!</i>	downstream gene variant, upstream gene variant	202	ENST00000432035 ?	ENSP00000455944	4
C3orf84 <i>e!</i>	downstream gene variant, upstream gene variant	166	ENST00000545770 NM_001080528.2	ENSP00000454903	4
CCDC36 <i>e!</i>	upstream gene variant	3314	ENST00000438782 ?	ENSP00000391788	1
CCDC36 <i>e!</i>	upstream gene variant	3927	ENST00000452691 NM_001135197.1	ENSP00000407837	1
CCDC36 <i>e!</i>	upstream gene variant	3937	ENST00000366429 ?	ENSP00000403700	1
CCDC36 <i>e!</i>	upstream gene variant	4044	ENST00000493870 ?	?	1
CCDC36 <i>e!</i>	upstream gene variant	2855	ENST00000296449 NM_178173.3	ENSP00000296449	1
CCDC71 <i>e!</i>	downstream gene variant, upstream gene variant	3339	ENST00000321895 NM_022903.3	ENSP00000319006	3
DALRD3 <i>e!</i>	upstream gene variant	4386	ENST00000492585 ?	?	1
DALRD3 <i>e!</i>	upstream gene variant	4384	ENST00000496568 ?	ENSP00000485305	1
IMPDH2 <i>e!</i>	upstream gene variant	1093	ENST00000442157 ?	ENSP00000403502	2
IMPDH2 <i>e!</i>	upstream gene variant	1998	ENST00000429182 ?	ENSP00000393525	1
IMPDH2 <i>e!</i>	upstream gene variant	2329	ENST00000462980 ?	?	1
IMPDH2 <i>e!</i>	upstream gene variant	2344	ENST00000491610 ?	?	1
IMPDH2 <i>e!</i>	upstream gene variant	1701	ENST00000472328 ?	?	1
IMPDH2 <i>e!</i>	downstream gene variant, upstream gene variant	1063	ENST00000496837 ?	?	3
IMPDH2 <i>e!</i>	upstream gene variant	1434	ENST00000484872 ?	?	1
IMPDH2 <i>e!</i>	downstream gene variant, upstream gene variant	1034	ENST00000485500 ?	?	3
IMPDH2 <i>e!</i>	upstream gene variant	1839	ENST00000463903 ?	?	1
IMPDH2 <i>e!</i>	upstream gene variant	1262	ENST00000481274 ?	?	1
IMPDH2 <i>e!</i>	upstream gene variant	1081	ENST00000326739 NM_000884.2	ENSP00000321584	2
IMPDH2 <i>e!</i>	upstream gene variant	1184	ENST00000466147 ?	?	1

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

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

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

QRICH1 <i>e!</i>	downstream gene variant, upstream gene variant	187	ENST00000498392 ?	?	4
QRICH1 <i>e!</i>	downstream gene variant, upstream gene variant	1059	ENST00000357496 NM_017730.2	ENSP00000350094	3
QRICH1 <i>e!</i>	downstream gene variant	3698	ENST00000489642 ?	?	1
QRICH1 <i>e!</i>	downstream gene variant, upstream gene variant	1510	ENST00000424300 ?	ENSP00000412890	3
RN75L182P <i>e!</i>	upstream gene variant, downstream gene variant	3852	ENST00000496499 ?	?	2
RP11-694115.7 <i>e!</i>	downstream gene variant	3163	ENST00000603877 ?	?	1
RP13-131K19.1 <i>e!</i>	downstream gene variant	143	ENST00000415982 ?	?	1
RP13-131K19.1 <i>e!</i>	downstream gene variant	157	ENST00000429681 ?	?	1
RP13-131K19.2 <i>e!</i>	upstream gene variant	4960	ENST00000452042 ?	?	1
RP13-131K19.6 <i>e!</i>	upstream gene variant, downstream gene variant	765	ENST00000607245 ?	?	3
SLC25A20 <i>e!</i>	downstream gene variant	1228	ENST00000430379 ?	ENSP00000388986	1
SLC25A20 <i>e!</i>	downstream gene variant	1233	ENST00000319017 NM_000387.5	ENSP00000326305	1
SLC25A20 <i>e!</i>	downstream gene variant	1689	ENST00000440964 ?	ENSP00000388563	1
SLC25A20 <i>e!</i>	downstream gene variant	1538	ENST00000479050 ?	?	1
USP19 <i>e!</i>	downstream gene variant, upstream gene variant	1062	ENST00000398896 ?	ENSP00000381870	2
USP19 <i>e!</i>	downstream gene variant	4367	ENST00000417901 NM_001199161.1	ENSP00000395260	1
USP19 <i>e!</i>	downstream gene variant	4363	ENST00000398898 ?	ENSP00000381872	1
USP19 <i>e!</i>	downstream gene variant	4990	ENST00000398892 ?	ENSP00000381867	1
USP19 <i>e!</i>	downstream gene variant	4992	ENST00000398888 NM_006677.2	ENSP00000381863	1
USP19 <i>e!</i>	upstream gene variant	1329	ENST00000464931 ?	?	1
USP19 <i>e!</i>	upstream gene variant	1252	ENST00000480163 ?	?	1
USP19 <i>e!</i>	downstream gene variant	4448	ENST00000453664 NM_001199162.1	ENSP00000400090	1
USP19 <i>e!</i>	upstream gene variant	1682	ENST00000479073 ?	ENSP00000428539	1
WDR6 <i>e!</i>	upstream gene variant	4354	ENST00000615452 ?	ENSP00000482289	1
WDR6 <i>e!</i>	upstream gene variant	117	ENST00000452875 ?	ENSP00000414157	1
WDR6 <i>e!</i>	upstream gene variant	152	ENST00000627177 ?	ENSP00000487056	1
WDR6 <i>e!</i>	upstream gene variant	123	ENST00000471162 ?	?	1
WDR6 <i>e!</i>	upstream gene variant	113	ENST00000438660 ?	ENSP00000387692	1
WDR6 <i>e!</i>	upstream gene variant	331	ENST00000419837 ?	ENSP00000390740	1
WDR6 <i>e!</i>	upstream gene variant	328	ENST00000472878 ?	?	1
WDR6 <i>e!</i>	upstream gene variant	82	ENST00000488572 ?	?	1
WDR6 <i>e!</i>	upstream gene variant	73	ENST00000429900 ?	ENSP00000477418	1
WDR6 <i>e!</i>	upstream gene variant	113	ENST00000608424 ?	ENSP00000477389	1
WDR6 <i>e!</i>	upstream gene variant	345	ENST00000461687 ?	?	1
WDR6 <i>e!</i>	upstream gene variant	113	ENST00000473238 ?	?	1
WDR6 <i>e!</i>	upstream gene variant	123	ENST00000420783 ?	ENSP00000395993	1
WDR6 <i>e!</i>	upstream gene variant	110	ENST00000462064 ?	?	1

WDR6 <i>e!</i>	upstream gene variant	116	ENST00000415265 ?	ENSP00000412195 1
WDR6 <i>e!</i>	upstream gene variant	328	ENST00000448293 ?	ENSP00000413432 1
WDR6 <i>e!</i>	upstream gene variant	786	ENST00000491365 ?	ENSP00000477463 1
WDR6 <i>e!</i>	upstream gene variant	781	ENST00000489427 ?	ENSP00000477087 1
WDR6 <i>e!</i>	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 <i>e!</i>	ENST00000545770 <i>e!</i>	NM_001080528.2	ENSP00000454903 <i>e!</i>	P	ccA/ccG	1
C3orf84 <i>e!</i>	ENST00000432035 <i>e!</i>	?	ENSP00000455944 <i>e!</i>	P	ccA/ccG	1
QARS <i>e!</i>	ENST00000306125 <i>e!</i>	NM_005051.2	ENSP00000307567 <i>e!</i>	H	caT/caC	1
QARS <i>e!</i>	ENST00000414533 <i>e!</i>	NM_001272073.1	ENSP00000390015 <i>e!</i>	H	caT/caC	1
QARS <i>e!</i>	ENST00000452739 <i>e!</i>	?	ENSP00000392850 <i>e!</i>	H	caT/caC	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
KLHDC8B <i>e!</i>	ENST00000332780 <i>e!</i>	NM_173546.2	ENSP00000327468 <i>e!</i>	1
KLHDC8B <i>e!</i>	ENST00000462582 <i>e!</i>	?	?	1

Intron variant

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

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

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

QRICH1 <i>e!</i>	ENST00000498440 <i>e!</i>	?	?	3
QRICH1 <i>e!</i>	ENST00000424300 <i>e!</i>	?	ENSP00000412890 <i>e!</i>	11
QRICH1 <i>e!</i>	ENST00000395443 <i>e!</i>	NM_198880.1	ENSP00000378830 <i>e!</i>	11
QRICH1 <i>e!</i>	ENST00000479449 <i>e!</i>	?	?	5
QRICH1 <i>e!</i>	ENST00000357496 <i>e!</i>	NM_017730.2	ENSP00000350094 <i>e!</i>	11
QRICH1 <i>e!</i>	ENST00000489642 <i>e!</i>	?	?	3
RP11-694115.7 <i>e!</i>	ENST00000603877 <i>e!</i>	?	?	3
SLC25A20 <i>e!</i>	ENST00000440964 <i>e!</i>	?	ENSP00000388563 <i>e!</i>	3
SLC25A20 <i>e!</i>	ENST00000319017 <i>e!</i>	NM_000387.5	ENSP00000326305 <i>e!</i>	3
SLC25A20 <i>e!</i>	ENST00000430379 <i>e!</i>	?	ENSP00000388986 <i>e!</i>	3

3'-UTR variant

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

5'-UTR variant

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

Non-coding exon variant

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

QARS <i>e!</i>	ENST00000464962 <i>e!</i>	?	1
QARS <i>e!</i>	ENST00000470619 <i>e!</i>	?	1
QARS <i>e!</i>	ENST00000479495 <i>e!</i>	?	1
QARS <i>e!</i>	ENST00000482261 <i>e!</i>	?	1
QRICH1 <i>e!</i>	ENST00000498392 <i>e!</i>	?	1
QRICH1 <i>e!</i>	ENST00000489642 <i>e!</i>	?	1
USP19 <i>e!</i>	ENST00000465902 <i>e!</i>	?	1
USP19 <i>e!</i>	ENST00000488993 <i>e!</i>	?	1
USP19 <i>e!</i>	ENST00000491859 <i>e!</i>	?	1

