UniProtKB - Q9NZ71 (RTEL1_HUMAN)

Protein **Regulator of telomere elongation helicase 1**

Gene **RTEL1**

Organism *Homo sapiens (Human)*

Status Reviewed - Annotation score: ●●●●● - Experimental evidence at protein level

Function

ATP-dependent DNA helicase implicated in telomere-length regulation, DNA repair and the maintenance of genomic stability. Acts as an anti-recombinase to counteract toxic recombination and limit crossover during meiosis. Regulates meiotic recombination and crossover homeostasis by physically dissociating strand invasion events and thereby promotes noncrossover repair by meiotic synthesis dependent strand annealing (SDSA) as well as disassembly of D loop recombination intermediates. Also disassembles T loops and prevents telomere fragility by counteracting telomeric G4-DNA structures, which together ensure the dynamics and stability of the telomere. Evidence: UniRule annotation Evidence: 3 Publications

Miscellaneous

Amplified in gastric tumors.

Catalytic activity

$ATP + H_2O = ADP + phosphate.$ Evidence: UniRule annotation

Sites

Feature key	Position(s)	Description	Graphical view
Metal binding	145	Iron-sulfur (4Fe-4S) Evidence: UniRule annotation	
Metal binding	163	Iron-sulfur (4Fe-4S) Evidence: UniRule annotation	
Metal binding	172	Iron-sulfur (4Fe-4S) Evidence: UniRule annotation	
Metal binding	207	Iron-sulfur (4Fe-4S) Evidence: UniRule annotation	

Regions

Feature key	Position(s)	Description	Graphical view
Nucleotide binding	42 – 49	ATP Evidence: Curated	

GO - Molecular function

- 4 iron, 4 sulfur cluster binding Evidence: Source: UniProtKB-KW
- ATP binding Evidence: Source: UniProtKB
- ATP-dependent DNA helicase activity Evidence: Source: UniProtKB
- DNA binding Evidence: Source: UniProtKB-KW
- metal ion binding Evidence: Source: UniProtKB-KW

GO - Biological process

- DNA duplex unwinding Evidence: Source: BHF-UCL
- DNA repair Evidence: Source: UniProtKB-KW
- mitotic telomere maintenance via semi-conservative replication Evidence: Source: BHF-UCL
- negative regulation of DNA recombination Evidence: Source: BHF-UCL
- negative regulation of t-circle formation Evidence: Source: BHF-UCL
- negative regulation of telomere maintenance in response to DNA damage Evidence: Source: BHF-UCL
- positive regulation of telomere capping Evidence: Source: BHF-UCL
- positive regulation of telomere maintenance Evidence: Source: BHF-UCL
- positive regulation of telomere maintenance via telomere lengthening Evidence: Source: BHF-UCL
- positive regulation of telomeric loop disassembly Evidence: Source: BHF-UCL
- regulation of double-strand break repair via homologous recombination Evidence: Source: UniProtKB
- replication fork processing Evidence: Source: BHF-UCL
- strand displacement Evidence: Source: BHF-UCL
- telomere maintenance Evidence: Source: UniProtKB
- telomere maintenance in response to DNA damage Evidence: Source: BHF-UCL
- telomeric loop disassembly Evidence: Source: BHF-UCL

Keywords

Molecular function	DNA-binding, Helicase, Hydrolase
Biological process	DNA damage, DNA repair
Ligand	4Fe-4S, ATP-binding, Iron, Iron-sulfur, Metal-binding, Nucleotide-binding

Enzyme and pathway databases

Reactome	R-HSA-2564830 Cytosolic iron-sulfur cluster assembly R-HSA-5693554 Resolution of D-loop Structures through Synthesis-Dependent Strand Annealing (SDSA)

Names & Taxonomy

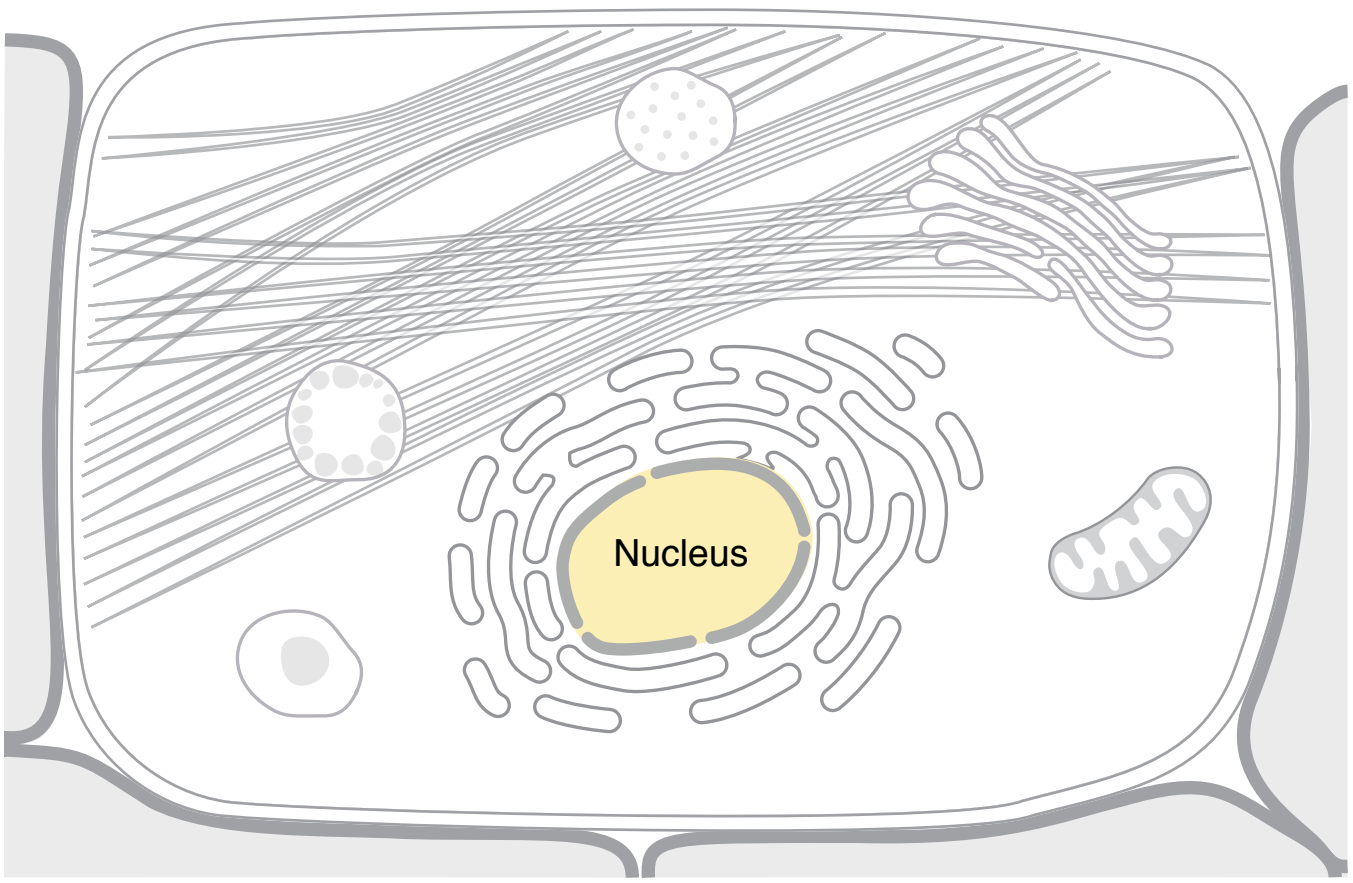
Protein names	*Recommended name:* **Regulator of telomere elongation helicase 1** Evidence: UniRule annotation (EC:3.6.4.12 Evidence: UniRule annotation) *Alternative name(s):* • Novel helicase-like
Gene names	*Name:*RTEL1 Evidence: UniRule annotation Synonyms:C20orf41, KIAA1088, NHL
Organism	Homo sapiens (Human)
Taxonomic identifier	9606 [NCBI]
Taxonomic lineage	Eukaryota › Metazoa › Chordata › Craniata › Vertebrata › Euteleostomi › Mammalia › Eutheria › Euarchontoglires › Primates › Haplorrhini › Catarrhini › Hominidae › Homo
Proteomes	UP000005640 Component: Chromosome 20

Organism-specific databases

EuPathDB	HostDB:ENSG00000258366.7
HGNC	HGNC:15888 RTEL1

MIM	608833 gene
neXtProt	NX_Q9NZ71

Subcellular location



Graphics by Christian Stolte; Source: COMPARTMENTS

Manual annotation Automatic computational assertion

UniProt annotation GO - Cellular component

Nucleus

Nucleus Evidence: UniRule annotation

Note: Colocalizes with PCNA within the replication foci in S-phase cells. Evidence: UniRule annotation

Keywords - Cellular component

Nucleus

Involvement in disease

Dyskeratosis congenita, autosomal recessive, 5 (DKCB5) Evidence: 5 Publications

The disease is caused by mutations affecting the gene represented in this entry. RTEL1 mutations have also been found in patients with a dyskeratosis congenita-like phenotype consisting of one feature of dyskeratosis congenita and short telomeres, in the absence of the typical DKC diagnostic triad (PubMed:23329068). Evidence: 1 Publication

Disease description: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. DKCB5 is characterized by onset of bone marrow failure and immunodeficiency in early childhood. Most patients also have growth and developmental delay and cerebellar hypoplasia, consistent with a clinical diagnosis of Hoyeraal-Hreidarsson syndrome.
See also OMIM:615190

Feature key	Position(s)	Description	Graphical view
Natural variant (VAR_069714)	251	E → K in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123019	
Natural variant (VAR_069715)	492	M → I in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 2 Publications Corresponds to variant dbSNP:rs370343781	
Natural variant (VAR_069716)	591	E → D in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123051	
Natural variant (VAR_069719)	699	I → M in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123048	
Natural variant (VAR_069720)	710	L → R in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication	
Natural variant (VAR_069721)	739	G → V in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123016	
Natural variant (VAR_069722)	745	V → M in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant	

		dbSNP:rs398123049	
Natural variant (VAR_069725)	897	K → E in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication	
Natural variant (VAR_069727)	957	R → W in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123018	
Natural variant (VAR_069728)	964	F → L in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication	
Isoform 5 (identifier: **Q9NZ71-5**)			
Natural variant	489	C → R in DKCB5, severe form consistent with Hoyeraal-Hreidarsson syndrome.	
Natural variant	509	R → H in DKCB5, abolishes activity.	
Isoform 1 (identifier: **Q9NZ71-2**)			
Natural variant	1244	C → R in DKCB5, severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: Curated	
Isoform 6 (identifier: **Q9NZ71-6**)			
Natural variant	1244	C → R in DKCB5, severe form consistent with Hoyeraal-Hreidarsson syndrome.	
Natural variant	1264	R → H in DKCB5, abolishes activity.	

Dyskeratosis congenita, autosomal dominant, 4 (DKCA4) Evidence: 1 Publication

The disease is caused by mutations affecting the gene represented in this entry.
Disease description: A rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy.
See also OMIM:615190

Feature key	Position(s)	Description	Graphical view
Natural variant (VAR_069717)	621	A → T in DKCA4. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123052	

Pulmonary fibrosis, and/or bone marrow failure, telomere-related, 3 (PFBMFT3)
Evidence: 1 Publication

The disease is caused by mutations affecting the gene represented in this entry.
Disease description: A disease associated with shortened telomeres. Pulmonary fibrosis is the most common manifestation. Other manifestations include aplastic anemia due to bone marrow failure, hepatic fibrosis, and increased cancer risk, particularly myelodysplastic syndrome and acute myeloid leukemia. Phenotype,

age at onset, and severity are determined by telomere length.
See also OMIM:616373

Feature key	Position(s)	Description	Graphical view
Natural variant (VAR_073795)	484	P → L in PFBMFT3. Evidence: 1 Publication Corresponds to variant dbSNP:rs786205700	
Natural variant (VAR_073796)	647	P → L in PFBMFT3. Evidence: 1 Publication	
Natural variant (VAR_073797)	1124	H → P in PFBMFT3. Evidence: 1 Publication Corresponds to variant dbSNP:rs786205702	

Mutagenesis

Feature key	Position(s)	Description	Graphical view
Mutagenesis	48	K → R: Abolishes ATPase activity. Evidence: 1 Publication	

Keywords - Disease
Disease mutation, Dyskeratosis congenita

Organism-specific databases

DisGeNET	51750
MalaCards	RTEL1
MIM	615190 phenotype 616373 phenotype
OpenTargets	ENSG00000258366
Orphanet	1775 Dyskeratosis congenita 3322 Hoyeraal-Hreidarsson syndrome
PharmGKB	PA134915625

Polymorphism and mutation databases

BioMuta	RTEL1
DMDM	229462743

PTM / Processing

Molecule processing

Feature key	Position(s)	Description	Graphical view
Chain (PRO_0000101985)	1 – 1219	Regulator of telomere elongation helicase 1	

Proteomic databases

EPD	Q9NZ71
MaxQB	Q9NZ71
PaxDb	Q9NZ71
PeptideAtlas	Q9NZ71
PRIDE	Q9NZ71

ProteomicsDB	83330 83331 [Q9NZ71-2] 83332 [Q9NZ71-4] 83333 [Q9NZ71-5] 83334 [Q9NZ71-6] 83335 [Q9NZ71-7] 83336 [Q9NZ71-8] 83337 [Q9NZ71-9]

PTM databases

iPTMnet	Q9NZ71
PhosphoSitePlus	Q9NZ71

Expression

Gene expression databases

Bgee	ENSG00000258366
ExpressionAtlas	Q9NZ71 baseline and differential
Genevisible	Q9NZ71 HS

Organism-specific databases

HPA	HPA020622 HPA067329 HPA078328

Interaction

Subunit structure

Interacts with TERF1. Interacts (via PIP-box) with PCNA; the interaction is direct and essential for suppressing telomere fragility. Interacts with MMS19; the interaction mediates the association of RTEL1 with the cytosolic iron-sulfur protein assembly (CIA) complex. Evidence: UniRule annotation

Evidence: 2 Publications

Protein-protein interaction databases

BioGrid	119711, 50 interactors
IntAct	Q9NZ71, 10 interactors
STRING	9606.ENSP00000359035

Structure

3D structure databases

ProteinModelPortal	Q9NZ71
ModBase	Search...
MobiDB	Search...

Family & Domains

Domains and Repeats

Feature key	Position(s)	Description	Graphical view
		Helicase ATP-binding	

Domain	7 – 296	Evidence: UniRule annotation	

Motif

Feature key	Position(s)	Description	Graphical view
Motif	151 – 167	Nuclear localization signal Evidence: UniRule annotation	
Motif	250 – 253	DEAH box	
Motif	871 – 877	Nuclear localization signal Evidence: UniRule annotation	
Motif	1178 – 1185	PIP-box	

Domain

The PIP-box (PCNA interacting peptide) motif mediates the interaction with PCNA and localization to replication foci. Evidence: UniRule annotation

Sequence similarities

Belongs to the helicase family. RAD3/XPD subfamily. Evidence: UniRule annotation

Phylogenomic databases

eggNOG	KOG1132 Eukaryota COG1199 LUCA
GeneTree	ENSGT00530000063199
HOVERGEN	HBG108423
InParanoid	Q9NZ71
KO	K11136
OrthoDB	EOG091G035L
PhylomeDB	Q9NZ71

Family and domain databases

HAMAP	MF_03065 RTEL1, 1 hit
InterPro	View protein in InterPro IPR006555 ATP-dep_Helicase_C IPR010614 DEAD_2 IPR014013 Helic_SF1/SF2_ATP-bd_DinG/Rad3 IPR006554 Helicase-like_DEXD_c2 IPR027417 P-loop_NTPase IPR013020 Rad3/Chl1-like IPR030845 RTEL1
Pfam	View protein in Pfam PF06733 DEAD_2, 1 hit PF13307 Helicase_C_2, 1 hit
SMART	View protein in SMART SM00488 DEXDc2, 1 hit SM00491 HELICc2, 1 hit
SUPFAM	SSF52540 SSF52540, 8 hits
TIGRFAMs	TIGR00604 rad3, 1 hit

PROSITE	View protein in PROSITE PS51193 HELICASE_ATP_BIND_2, 1 hit

Sequences (8)

Sequence status: Complete.
This entry describes **8** isoforms produced by **alternative splicing**.

Note: Additional isoforms seem to exist.

Isoform 2 (identifier: **Q9NZ71-1**) [UniParc]
This isoform has been chosen as the 'canonical' sequence. All positional information in this entry refers to t
This is also the sequence that appears in the downloadable versions of the entry.
« Hide

```
        10         20         30         40         50
MPKIVLNGVT VDFPFQPYKC QQEYMTKVLE CLQQKVNGIL ESPTGTGKTL
        60         70         80         90        100
CLLCTTLAWR EHLRDGISAR KIAERAQGEL FPDRALSSWG NAAAAAGDPI
       110        120        130        140        150
ACYTDIPKII YASRTHSQLT QVINELRNTS YRPKVCVLGS REQLCIHPEV
       160        170        180        190        200
KKQESNHLQI HLCRKKVASR SCHFYNNVEE KSLEQELASP ILDIEDLVKS
       210        220        230        240        250
GSKHRVCPYY LSRNLKQQAD IIFMPYNYLL DAKSRRAHNI DLKGTVVIFD
       260        270        280        290        300
EAHNVEKMCE ESASFDLTPH DLASGLDVID QVLEEQTKAA QQGEPHPEFS
       310        320        330        340        350
ADSPSPGLNM ELEDIAKLKM ILLRLEGAID AVELPGDDSG VTKPGSYIFE
       360        370        380        390        400
LFAEAQITFQ TKGCILDSLD QIIQHLAGRA GVFTNTAGLQ KLADIIQIVF
       410        420        430        440        450
SVDPSEGSPG SPAGLGALQS YKVHIHPDAG HRRTAQRSDA WSTTAARKRG
       460        470        480        490        500
KVLSYWCFSP GHSMHELVRQ GVRSLILTSG TLAPVSSFAL EMQIPFPVCL
       510        520        530        540        550
ENPHIIDKHQ IWVGVVPRGP DGAQLSSAFD RRFSEECLSS LGKALGNIAR
       560        570        580        590        600
VVPYGLLIFF PSYPVMEKSL EFWRARDLAR KMEALKPLFV EPRSKGSFSE
       610        620        630        640        650
TISAYYARVA APGSTGATFL AVCRGKASEG LDFSDTNGRG VIVTGLPYPP
       660        670        680        690        700
RMDPRVVLKM QFLDEMKGQG GAGGQFLSGQ EWYRQQASRA VNQAIGRVIR
       710        720        730        740        750
HRQDYGAVFL CDHRFAFADA RAQLPSWVRP HVRVYDNFGH VIRDVAQFFR
       760        770        780        790        800
VAERTMPAPA PRATAPSVRG EDAVSEAKSP GPFFSTRKAK SLDLHVPSLK
       810        820        830        840        850
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QRSSGSPAAG DPESSLCVEY EQEPVPARQR PRGLLAALEH SEQRAGSPGE
       860        870        880        890        900
EQAHSCSTLS LLSEKRPAEE PRGGRKKIRL VSHPEEPVAG AQTDRAKLFM
       910        920        930        940        950
VAVKQELSQA NFATFTQALQ DYKGSDDFAA LAACLGPLFA EDPKKHNLLQ
       960        970        980        990       1000
GFYQFVRPHH KQQFEEVCIQ LTGRGCGYRP EHSIPRRQRA QPVLDPTGRT
      1010       1020       1030       1040       1050
APDPKLTVST AAAQQLDPQE HLNQGRPHLS PRPPPTGDPG SQPQWGSGVP
      1060       1070       1080       1090       1100
RAGKQGQHAV SAYLADARRA LGSAGCSQLL AALTAYKQDD DLDKVLAVLA
      1110       1120       1130       1140       1150
ALTTAKPEDF PLLHRFSMFV RPHHKQRFSQ TCTDLTGRPY PGMEPPGPQE
      1160       1170       1180       1190       1200
ERLAVPPVLT HRAPQPGPSR SEKTGKTQSK ISSFLRQRPA GTVGAGGEDA
      1210
GPSQSSGPPH GPAASEWGL
```

Length: 1,219
Mass (Da): 133,683
Last modified: May 5, 2009 - v2
Checksum: 28DFCFCC48BC0055

Isoform 1 (identifier: **Q9NZ71-2**) [UniParc]
The sequence of this isoform differs from the canonical sequence as follows:
1219-1219: L → EPHGRDIAGQ...PLLQRPLRGA

Note: Variant in position: 1264:R->H (in DKCB5), abolishes activity. Evidence: Curated
Show »

Length: 1,400
Mass (Da): 152,374
Checksum: F3F2BB93D48ED3D9

Isoform 4 (identifier: **Q9NZ71-4**) [UniParc]
The sequence of this isoform differs from the canonical sequence as follows:
999-1023: RTAPDPKLTVSTAAAQQLDPQEHLN → NFPDALDQLCGSTSLHQEERRRIPS
1024-1219: Missing.

Note: No experimental confirmation available.
Show »

Length: 1,023
Mass (Da): 113,184
Checksum: 43650D4EC91B6DEA

Isoform 5 (identifier: **Q9NZ71-5**) [UniParc]

The sequence of this isoform differs from the canonical sequence as follows:

1-755: Missing.

1219-1219: L → EPHGRDIAGQ...VMQVFWPEPQ

Note: No experimental confirmation available.

Show »

Length: 545

Mass (Da): 58,545

Checksum: A08763FAE15AE678

Isoform 6 (identifier: **Q9NZ71-6**) [UniParc]

The sequence of this isoform differs from the canonical sequence as follows:

1219-1219: L → EPHGRDIAGQ...VMQVFWPEPQ

Note: No experimental confirmation available.

Show »

Length: 1,300

Mass (Da): 142,367

Checksum: E2A1CD6CC3211479

Isoform 7 (identifier: **Q9NZ71-7**) [UniParc]

The sequence of this isoform differs from the canonical sequence as follows:

131-131: Y → YRSRCRATLWVLETAPPRPTVLSPT

Note: No experimental confirmation available.

Show »

Length: 1,243

Mass (Da): 136,373

Checksum: 5AFDE395097DDC14

Isoform 8 (identifier: **Q9NZ71-8**) [UniParc]

The sequence of this isoform differs from the canonical sequence as follows:

998-1219: GRTAPDPKLT...HGPAASEWGL → ERRRIPS

Note: No experimental confirmation available.

Show »

Length: 1,004

Mass (Da): 111,170

Checksum: D3F25736F4CD034A

Isoform 9 (identifier: **Q9NZ71-9**) [UniParc]

The sequence of this isoform differs from the canonical sequence as follows:

1-223: Missing.

Note: No experimental confirmation available.

Show »

Length: 996
Mass (Da): 108,457
Checksum: 6CD391EF1A61C675

Experimental Info

Feature key	Position(s)	Description	Graphical view
Sequence conflict	41	E → G in BAG63785 (PubMed:14702039). Evidence: Curated	
Sequence conflict	48	K → R in BAG61337 (PubMed:14702039). Evidence: Curated	
Sequence conflict	845	A → V in BAG63785 (PubMed:14702039). Evidence: Curated	
Sequence conflict	986	R → Q in BAG61337 (PubMed:14702039). Evidence: Curated	
Isoform 1 (identifier: **Q9NZ71-2**)			
Sequence conflict	1352	C → R in BAA83040 (PubMed:10470851). Evidence: Curated	

Natural variant

Feature key	Position(s)	Description	Graphical view
Natural variant (VAR_054970)	124	N → S Evidence: 1 Publication Corresponds to variant dbSNP:rs3848668	
Natural variant (VAR_069714)	251	E → K in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123019	
Natural variant (VAR_073795)	484	P → L in PFBMFT3. Evidence: 1 Publication Corresponds to variant dbSNP:rs786205700	
Natural variant (VAR_069715)	492	M → I in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 2 Publications Corresponds to variant dbSNP:rs370343781	
Natural variant (VAR_069716)	591	E → D in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123051	
Natural variant (VAR_069717)	621	A → T in DKCA4. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123052	

Natural variant (VAR_073796)	647	P → L in PFBMFT3. Evidence: 1 Publication
Natural variant (VAR_069718)	684	R → Q Evidence: 1 Publication Corresponds to variant dbSNP:rs35640778
Natural variant (VAR_069719)	699	I → M in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123048
Natural variant (VAR_069720)	710	L → R in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication
Natural variant (VAR_069721)	739	G → V in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123016
Natural variant (VAR_069722)	745	V → M in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123049
Natural variant (VAR_069723)	829	Q → P Evidence: 1 Publication
Natural variant (VAR_069724)	849	G → D Evidence: 1 Publication Corresponds to variant dbSNP:rs190887884
Natural variant (VAR_069725)	897	K → E in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication
Natural variant (VAR_069726)	929	A → T Evidence: 2 Publications Corresponds to variant dbSNP:rs61736615
Natural variant (VAR_069727)	957	R → W in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication Corresponds to variant dbSNP:rs398123018
Natural variant (VAR_069728)	964	F → L in DKCB5; severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: 1 Publication
Natural variant (VAR_069729)	1034	P → H Evidence: 1 Publication Corresponds to variant dbSNP:rs115610405
Natural variant (VAR_054971)	1042	Q → H Evidence: 3 Publications Corresponds to variant dbSNP:rs3208008
Natural variant (VAR_069730)	1059	A → T Evidence: 1 Publication Corresponds to variant dbSNP:rs115303435
Natural variant (VAR_073797)	1124	H → P in PFBMFT3. Evidence: 1 Publication Corresponds to variant dbSNP:rs786205702

Isoform 5 (identifier: **Q9NZ71-5**)			
Natural variant	489	C → R in DKCB5, severe form consistent with Hoyeraal-Hreidarsson syndrome.	
Natural variant	509	R → H in DKCB5, abolishes activity.	
Isoform 1 (identifier: **Q9NZ71-2**)			
Natural variant	1244	C → R in DKCB5, severe form consistent with Hoyeraal-Hreidarsson syndrome. Evidence: Curated	
Isoform 6 (identifier: **Q9NZ71-6**)			
Natural variant	1244	C → R in DKCB5, severe form consistent with Hoyeraal-Hreidarsson syndrome.	
Natural variant	1264	R → H in DKCB5, abolishes activity.	

Alternative sequence

Feature key	Position(s)	Description	Graphical view
Alternative sequence (VSP_017093)	1 – 755	Missing in isoform 5. Evidence: 1 Publication	
Alternative sequence (VSP_036937)	1 – 223	Missing in isoform 9. Evidence: 1 Publication	
Alternative sequence (VSP_036938)	131	Y → YRSRCRATLWVLETAPPRPT VLSPT in isoform 7. Evidence: 1 Publication	
Alternative sequence (VSP_036939)	998 – 1219	GRTAP...SEWGL → ERRRIPS in isoform 8. Evidence: 1 Publication	
Alternative sequence (VSP_007076)	999 – 1023	RTAPD...QEHLN → NFPDALDQLCGSTSLHQEER RRIPS in isoform 4. Evidence: 1 Publication	
Alternative sequence (VSP_007077)	1024 – 1219	Missing in isoform 4. Evidence: 1 Publication	
Alternative sequence (VSP_036940)	1219	L → EPHGRDIAGQQATGAPGGPL SAGCVCQGCGAEDVVPFQCP ACDFQRCQACWQRHLQASRM CPACHTASRKQSVMQVFWPE PHKDHEGAGGARPVAAVPGV GAACPAAGAGCTRSGRNTHL PLAGRRDRGAAGVCPVPPRH LCAAAVPPRQPHDVWPVSTA PLHAVLELPGALPLLQRPLR GA in isoform 1. Evidence: 2 Publications	
Alternative sequence (VSP_017094)	1219	L → EPHGRDIAGQQATGAPGGPL SAGCVCQGCGAEDVVPFQCP ACDFQRCQACWQRHLQASRM CPACHTASRKQSVMQVFWPE PQ in isoform 5 and isoform 6. Evidence: 1 Publication	

Sequence databases

Select the link destinations:
EMBL

AF217795 mRNA Translation: AAF33687.1
AF217796 Genomic DNA Translation: AAF35243.1
AB029011 mRNA Translation: BAA83040.3

GenBank DDBJ	AK000485 mRNA Translation: BAA91197.1 AK302508 mRNA Translation: BAG63785.1 AK299332 mRNA Translation: BAG61337.1 AK304798 mRNA Translation: BAG65548.1 AL353715 Genomic DNA No translation available. CH471077 Genomic DNA Translation: EAW75238.1 CH471077 Genomic DNA Translation: EAW75239.1 CH471077 Genomic DNA Translation: EAW75240.1 CH471077 Genomic DNA Translation: EAW75241.1 CH471077 Genomic DNA Translation: EAW75245.1 AL080127 mRNA Translation: CAB45725.1
CCDS	CCDS13530.3 [Q9NZ71-7] CCDS13531.1 [Q9NZ71-1] CCDS63331.1 [Q9NZ71-6] CCDS74751.1 [Q9NZ71-9]
PIR	T12516 T45294
RefSeq	NP_001269938.1, NM_001283009.1 [Q9NZ71-6] NP_001269939.1, NM_001283010.1 [Q9NZ71-9] NP_057518.1, NM_016434.3 [Q9NZ71-1] NP_116575.3, NM_032957.4 [Q9NZ71-7]
UniGene	Hs.745057

Genome annotation databases

Ensembl	ENST00000318100; ENSP00000322287; ENSG00000258366 [Q9NZ71-9] ENST00000360203; ENSP00000353332; ENSG00000258366 [Q9NZ71-6] ENST00000370018; ENSP00000359035; ENSG00000258366 [Q9NZ71-1] ENST00000482936; ENSP00000457868; ENSG00000258366 [Q9NZ71-8] ENST00000508582; ENSP00000424307; ENSG00000258366 [Q9NZ71-7]
GeneID	51750
KEGG	hsa:51750
UCSC	uc002yfu.3 human [Q9NZ71-1]

Keywords - Coding sequence diversity

Alternative splicing, Polymorphism

Similar proteins

90% Identity 50% Identity

Protein	Similar proteins		Species	Score	Length
Q9NZ71	UPI0007DBBE73		PANTR		1521
	Regulator of telomere elongation helicase 1		PANPA	●●○○○	1318
	Regulator of telomere elongation helicase 1		MACFA	●●●○○	1306
	Regulator of telomere elongation helicase 1		PONAB	●●●○○	1302
	UPI00053317EF		RHIRO		1301
	+31				
Q9NZ71-2	UPI0007326F6D		MACMU		1466

	Regulator of telomere elongation helicase 1		CHLSB	●●●●○	1406
	Regulator of telomere elongation helicase 1		HUMAN	●●●○○	1400
	Regulator of telomere elongation helicase 1		PAPAN	●●●○○	1306
	Regulator of telomere elongation helicase 1		MACNE	●●●○○	1305
	+21				
Q9NZ71-4	Regulator of telomere elongation helicase 1 (Fragment)		HUMAN	●○○○○	316
	Regulator of telomere elongation helicase 1		RHIBE	●●●○○	1277
	UPI00083C323E		RHIBE		1229
Q9NZ71-7	Regulator of telomere elongation helicase 1		PAPAN	●●●○○	1355
	Regulator of telomere elongation helicase 1		MACNE	●●●○○	1354
	Regulator of telomere elongation helicase 1		CERAT	●●●○○	1352
	Regulator of telomere elongation helicase 1		MACFA	●●●○○	1352
	Regulator of telomere elongation helicase 1		MANLE	●●●○○	1351
	+8				
Q9NZ71-9	Regulator of telomere elongation helicase 1		PAPAN	●●●○○	1355
	Regulator of telomere elongation helicase 1		MACNE	●●●○○	1354
	Regulator of telomere elongation helicase 1		CERAT	●●●○○	1352
	Regulator of telomere elongation helicase 1		MACFA	●●●○○	1352
	Regulator of telomere elongation helicase 1		MANLE	●●●○○	1351
	+8				

Entry information

Entry name	RTEL1_HUMAN	
Accession	Primary (citable) accession number: **Q9NZ71** Secondary accession number(s): A2A397 Q9Y4R6	
Entry history	Integrated into UniProtKB/Swiss-Prot:	March 28, 2003
	Last sequence update:	May 5, 2009
	Last modified:	July 18, 2018
	This is version 155 of the entry and version 2 of the sequence. See complete history.	
Entry status	Reviewed (UniProtKB/Swiss-Prot)	
Annotation program	Chordata Protein Annotation Program	
Disclaimer	Any medical or genetic information present in this entry is provided for research, educational and informational purposes only. It is not in any way intended to be used as a substitute for	

professional medical advice, diagnosis, treatment or care.

Miscellaneous

Keywords - Technical term
Complete proteome, Reference proteome

Documents

- Human chromosome 20
  Human chromosome 20: entries, gene names and cross-references to MIM
- Human entries with polymorphisms or disease mutations
  List of human entries with polymorphisms or disease mutations
- Human polymorphisms and disease mutations
  Index of human polymorphisms and disease mutations
- MIM cross-references
  Online Mendelian Inheritance in Man (MIM) cross-references in UniProtKB/Swiss-Prot
- SIMILARITY comments
  Index of protein domains and families

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EMBL European Molecular Biology Laboratory

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