Title: Dyskeratosis Congenita *GeneReview* Supplemental Material – Table 4
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Table 4. Selected *TERC* Pathogenic Variants

RNA Nucleotide Change (Alias [1])	Reference Sequences	Literature Reference [2]
r.-240delct	NR_001566.1	Field et al [2006]
(C-99G) [3]		Keith et al [2004]
r.2g>c (G2C)		Marrone et al [2007]
Contiguous gene deletion [4] (Δ1- 316)		Vulliamy et al [2004]
r.21c>u		Fogarty et al [2003]
r.28_34del7 (Δ28-34)		Xin et al [2007]
r.35c>u		Du et al [2009]
r.37a>g (A37G)		Vulliamy et al [2006]
r.48a>g (A48G)		Vulliamy et al [2006]
r.52_55delcuaa		Vulliamy et al [2006]
r.53_87del35		Marrone et al [2007]
r.58g>a (G58A)		Dokal & Vulliamy [2003]
r.72c>g		Dokal & Vulliamy [2003]
r.79delc		Vulliamy et al [2006]
r.96-97delcu		Vulliamy et al [2004]
r.98g>a (G98A)		Calado & Young [2008]
r.100u>a (T100A)		Du et al [2009]
c.110_113delgact		Walne & Dokal [2004]
c.107_108gc>ag		Vulliamy et al [2001]
r.116c>u (C116T)		Walne & Dokal [2004]
r.117a>c (A117C)		Ly et al [2005]
r.143g>a (G143A)		Vulliamy et al [2004]
r.178g>a (G178A)		Marrone et al [2007a]
r.180c>u (C180T)		Marrone et al [2007a]
r.204c>g		Fogarty et al [2003]

(C204G)		
r.216_229del14 (Del 216-229)		Vulliamy et al [2006]
r.228G>A (G228A)		Walne & Dokal [2004]
r.305g>a (G305A)		Fogarty et al [2003]
r.322g>a (G322A)		Fogarty et al [2003]
r.323c>u (C323T)		Calado & Young [2008]
(del 378 through 3' end of *TERC*)		Vulliamy et al [2004]
r.378_415del38		Dokal & Vulliamy [2003]
r.391_392delcc (Δ389-390)		Ly et al [2005]
r.408c>g		Vulliamy et al [2001]
r.408c>a		Marrone et al [2005]
r.410c>g		Vulliamy et al [2001]
r.450g>a		Walne & Dokal [2004]
(16u>c, 16 bp downstream of 3' transcript of *TERC*)		Fogarty et al [2003]
(821-bp deletion including 3' end of *TERC*)		Vulliamy et al [2001]

For this gene: the prefix "r." is used to indicate that a change is described at RNA level; numbering is relative to the transcription start site; nucleotides are designated by the bases (in lower case); bases are a (adenine), c (cytosine), g (guanine), and u (uracil).

Note on variant classification: Variants listed in the table have been provided by the author. *GeneReviews* staff have not independently verified the classification of variants.

Note on nomenclature: *GeneReviews* follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org). See Quick Reference for an explanation of nomenclature.

1. Variant designation that does not conform to current naming conventions

2. First literature reference given when possible

3. Promoter mutation in a individual with paroxysmal nocturnal hemoglobinuria

4. Deletion of 2980 bp extending from nucleotide 835 in the 3' UTR of *ACTRT3* (*ARPM1*), through the intergenic and TERC promoter sequences, to nucleotide 316 of *TERC*.

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