Title: Dyskeratosis Congenita *GeneReview* Supplemental Material – Table 6
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Table 6. Selected *TINF2* Pathogenic Variants

DNA Nucleotide Change (Alias [1])	Predicted Protein Change (Alias [1])	Reference Sequences	Literature Reference
c.805C>T	p.Gln269Ter	NM_001099274.1 NP_001092744.1	Sasa et al [2012]
c.811C>T	p.Gln271Ter		Sasa et al [2012]
(Ex6+234A>G)	(Lys280Glu) [2]		Savage et al [2008]
c.838A>T	p.Lys280Ter		Walne et al [2008] Sasa et al [2012]
c.839delA	p.Lys280ArgfsTer37 (Lys280ArgfsTer36)		
c.844C>T	p.Arg282Cys		
(Ex6+240C>A)	(Arg282Ser) [2]		Savage et al [2008]
c.847C>T	p.Pro283Ser		Walne et al [2008]
c.847C>G	p.Pro283Ala		
c.848C>A	p.Pro283His		
c.850A>G	p.Thr284Ala		
c.849dupC (849_850insC)	p.Thr284HisfsTer8		
c.860T>C	p.Leu287Pro		
c.862T>C	p.Phe288Leu [3]		Du et al [2009]
c.865CC>AG	p.Pro289Ser		Walne et al [2008]
c.871A>G	p.Arg291Gly		
c.892delC	p.Gln298ArgfsTer19		
c.706C>T	p.Pro236Ser [3]		
c.734C>A	p.Ser245Tyr [3]		
c.841G>A	p.Glu281Lys [3]		

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. Predicted from in silico analyses identifying putative exon splicing enhancers [Savage et al 2008]

3. Individual with bone marrow failure only

References

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Sasa G, Ribes-Zamora A, Nelson N, Bertuch A. Three novel truncating TINF2 mutations causing severe dyskeratosis congenita in early childhood. Clin Genet. 2012 81:470-8.

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Walne AJ, Vulliamy T, Beswick R, Kirwan M, Dokal I. TINF2 mutations result in very short telomeres: analysis of a large cohort of patients with dyskeratosis congenita and related bone marrow failure syndromes. Blood. 2008 Nov 1;112(9):3594-600.