Title: Dyskeratosis Congenita *GeneReview* Supplemental Material – Table 5
Author: Savage SA
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**Table 5. Selected *TERT* Allelic Variants**

| Class of Variant Allele | DNA Nucleotide Change (Alias [1]) | Predicted Protein Change | Disorder | Reference Sequences | Literature Reference |
|---|---|---|---|---|---|
| **Benign** | c.915G>A | p.Ala305Ala | Normal variant | NM_198253.2 NP_937983.2 | Vulliamy et al [2005] |
| | c.2097 C>T | p.Ala699Ala | Normal variant | | Vulliamy et al [2005], Yamaguchi et al [2005] |
| | c.2178 G>A | p.Thr726Thr | Normal variant | | Vulliamy et al [2005], Yamaguchi et al [2005] |
| | c.3039 C>T | p.His1013His | Normal variant | | Vulliamy et al [2005], Yamaguchi et al [2005] |
| **Pathogenic** | c.97C>T | p.Pro33Ser | Idiopathic pulmonary fibrosis | | Tsakiri et al [2007] |
| | c.164T>A | p.Leu55Gln | Idiopathic pulmonary fibrosis | | |
| | c.164T>A | p.Leu55Gln | Idiopathic pulmonary fibrosis | | Armanios et al [2007] |
| | c.112delC | p.Leu38TrpfsTer40 | Idiopathic pulmonary fibrosis | | Armanios et al [2007] |
| | c.430G>A | p.Val144Met | Idiopathic pulmonary fibrosis | | Tsakiri et al [2007] |
| | c.604G>A | p.Ala202Thr | Aplastic anemia | | Yamaguchi et al [2005] |
| | c.835G>A | p.Ala279Thr | Dyskeratosis congenita/ aplastic anemia | | Vulliamy et al [2005] |
| | c.1234C>T | p.His412Tyr | Aplastic anemia | | Yamaguchi et al [2005] |
| | c.1378_1380delCAG | p.441Gludel | Normal variant | | Yamaguchi et al [2005] |
| | c.1456C>T | p.Arg486Cys | Idiopathic pulmonary fibrosis | | Tsakiri et al [2007] |
| | c. 1710(G>T;G>C) [2] | p.Lys570Asn | Dyskeratosis congenita | | Xin et al [2007] |
| | c.1892G>A | p.Arg631Gln | Thrombocytopenia/ pulmonary fibrosis | | Kirwan et al [2008] |

| Class of Variant Allele | DNA Nucleotide Change (Alias [1]) | Predicted Protein Change | Disorder | Reference Sequences | Literature Reference |
|---|---|---|---|---|---|
| | c.2045G>A | p.Gly682Asp | Dyskeratosis congenita | | Xin et al [2007] |
| | c.2029G>T | p.Gly677Cys | Dyskeratosis congenita/ aplastic anemia | | Yamaguchi et al [2005] |
| | c.2080G>A | p.Val694Met | Aplastic anemia | | Yamaguchi et al [2005] |
| | c.2110C>T | p.Pro704Ser | Dyskeratosis congenita | | Du et al [2009] |
| | c.2147C>T | p.Ala716Val | Aplastic anemia | | Du et al [2009] |
| | c.2162C>G | p.Pro721Arg | Dyskeratosis congenita | | Vulliamy et al [2006] |
| | c.2177C>T | p.Thr726Met | Dyskeratosis congenita | | Xin et al [2007] |
| | c.2240delT | p.Val747AlafsTer20 | Idiopathic pulmonary fibrosis | | Tsakiri et al [2007] |
| | c.2315A>G | p.Tyr772Cys | Idiopathic AA | | Yamaguchi et al [2005] |
| | c.2431C>T | p.Arg811Cys | Autosomal recessive DC | | Marrone et al [2007] |
| | c.2537A>G | p.Tyr846Cys | Aplastic anemia | | Du et al [2009] |
| | c.259G>A | p.Arg865His | Idiopathic pulmonary fibrosis | | Tsakiri et al [2007] |
| | c.2628C>G | p.His876Gln | Aplastic anemia | | Du et al [2008] |
| | c.2701C>T | p.Arg901Trp | Autosomal recessive DC | | Marrone et al [2007] |
| | c.2706G>C | p.Lys902Asn | Dyskeratosis congenita | | Armanios et al [2007] |
| | c.2935C>T | p.Arg979Trp | Dyskeratosis congenita | | Vulliamy et al [2005], Savage et al [2006] |
| | c.3329C>T | p.Thr1110Met | Idiopathic pulmonary fibrosis | | Armanios et al [2007] |
| | c.3043T>C | p.Cys1015Arg | Aplastic anemia | | Du et al [2009] |
| | c.3184G>A | p.Ala1062Thr | Aplastic anemia | | Yamaguchi et al [2005] |
| | c.3268G>A | p.Val1090Met | Aplastic anemia | | Yamaguchi et al [2005] |
| | c.3404_3580del177 (3346_3522del) | | Idiopathic pulmonary fibrosis | | Tsakiri et al [2007] |
| | c.219+1G>A (IVS1+1G>A) | -- | Usual interstitial pneumonia | | Armanios et al [2007] |

| Class of Variant Allele | DNA Nucleotide Change (Alias [1]) | Predicted Protein Change | Disorder | Reference Sequences | Literature Reference |
|---|---|---|---|---|---|
| | c.2613-2A>G (IVS9-2 A>C) | -- | Idiopathic interstitial pneumonia | | Armanios et al [2007] |

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. Uncertainty of nucleotide change in parenthesis

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