

Table 2. Selected *DKC1* Pathogenic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences	Literature Reference
c.-142C>G	--	NM_001363.3 NP_001354.1	Walne & Dokal [2004]
c.5C>T	p.Ala2Val		Dokal & Vulliamy [2003]
c.29C>T	p.Pro10Leu		Marrone et al [2005]
c.91C>G	p.Gln31Glu		Vulliamy et al [2006]
c.91C>A	p.Gln31Lys		Heiss et al [1998]
c.106T>G	p.Phe36Val		Dokal & Vulliamy [2003]
c.109_111delCTT	p.Leu37del		Heiss et al [1998]
c.113T>C	p.Ile38Thr		Dokal & Vulliamy [2003]
c.115A>G	p.Lys39Glu		Heiss et al [1998]
c.119C>G	p.Pro40Arg		Dokal & Vulliamy [2003]
c.121G>A	p.Glu41Lys		Knight et al [1999]
c.127A>G	p.Lys43Glu		Dokal & Vulliamy [2003]
c.146C>T	p.Thr49Met		Knight et al [1999]
c.194G>C	p.Arg65Thr		Dokal & Vulliamy [2003]
c.196A>G	p.Thr66Ala		Marrone et al [2005]
c.200C>T	p.Thr67Ile		Knight et al [1999]
c.204C>A	p.His68Gln		Knight et al [2001]
c.361A>G	p.Ser121Gly		Du et al [2009]
c.472C>T	p.Arg158Trp		Dokal & Vulliamy [2003]
c.838A>C	p.Ser280Arg		Marrone et al [2005]
c.911G>A	p.Ser304Asn		Du et al [2009]
c.949C>T	p.Leu317Phe		Dokal & Vulliamy [2003]
c.941A>G	p.Lys314Arg		Marrone et al [2005]
c.949C>T	p.Leu317Phe		Du et al [2009]
c.949C>G	p.Leu317Val		Dokal & Vulliamy [2003]
c.961C>G	p.Leu321Val		Knight et al [2001]
c.965G>A	p.Arg322Gln		Dokal & Vulliamy [2003]
c.1049T>C	p.Met350Thr		Knight et al [2001]
c.1050G>A	p.Met350Ile		Dokal & Vulliamy [2003]
c.1058C>T	p.Ala353Val		Knight et al [2001]
c.1075G>A	p.Asp359Asn		Marrone et al [2005]
c.1150C>T	p.Pro384Ser		Dokal & Vulliamy [2003]
c.1151C>T	p.Pro384Leu		Knight et al [2001]

DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences	Literature Reference
c.1156G>A	p.Ala386Thr		Marrone et al [2005]
c.1193T>C	p.Leu398Pro		Dokal & Vulliamy [2003]
c.1205G>A	p.Gly402Glu		Heiss et al [1998]
c.1204G>A	p.Gly402Arg		Dokal & Vulliamy [2003]
c.1223C>T	p.Thr408Ile		Marrone et al [2005]
c.1226C>T	p.Pro409Leu		
c.1205G>A	p.Gly402Glu		Kirwan et al [2008]

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.

References

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