

Title: *TUBB4A*-Related Leukodystrophy *GeneReview* Table 3

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 3. *TUBB4A* Selected Pathogenic Variants

Amino Change	Nucleic Acid Change	Reference Sequence
p.Arg2Gly	c.4C>G ¹	NM_006087.3 NP_006078.2
p.Arg2Trp	4C>T ²	
p.Arg2Gln	c.5G>A ^{2,3}	
p.Arg156Leu	c.467G>T ⁴	
p.Thr178Arg	c.533C>G ³	
p.Thr178Met	c.533C>T ⁵	
p.Pro182Thr	c.544C>A ⁵	
p.His190Tyr	c.568C>T ⁶	
p.Cys239Phe	c.716G>T ^{2,7}	
p.Gly244Ser	c.730G>A ²	
p.Gly244Val	c.731G>T ²	
p.Gly244Asp	c.731G>A ⁵	
p.Asp249Asn	c.745G>A ^{2,3,7,8}	
p.Val255Ile	c.763G>A ⁹	
p.Arg262His	c.785G>A ^{3,7}	
p.Arg282Pro	c.845G>C ⁹	
p.Gln292Lys	c.874C>A ⁹	
p.Met300Ile	c.900C>A ¹⁰	
p.Met300Ile	c.900G>T ¹¹	
p.Ala314Val	c.941C>T ¹¹	
p.Met323Arg	c.968T>G ²	
p.Ala352Thr	c.1054G>A ²	
p.Cys354Tyr	c.1061G>A ²	
p.Ala364Asp	c.1091C>A ¹⁰	
p.Phe367Ile	c.1099T>A ²	
p.Phe367Leu	c.1099T>C ²	
p.Met388Val	c.1162A>G ³	

Amino Change	Nucleic Acid Change	Reference Sequence
p.Met388Thr	c.1163T>C ²	
p.Met388Ile	c.1164G>A ²	
p.Arg391His	c.1172G>A ⁹	
p.Phe394Cys	c.1181T>G ¹²	
p.Glu410Lys	c.1228G>A ³	

Note on variant classification: Variants listed in the table have been provided by the authors. *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. Hersheson et al [2013]
2. Hamilton et al [2014]
3. Miyatake et al [2014]
4. Purnell et al [2014]
5. Tonduti et al [2016]
6. Kancheva et al [2015]
7. Ferreira et al [2014]
8. Simons et al [2013]
9. Pizzino al [2014]
10. Pyle et al [2015]
11. Erro et al [2015a], Erro et al [2015b]
12. Carvalho et al [2015]

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