

Title: *TBC1D24*-Related Disorders *GeneReview* Table 2

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

Table 2. *TBC1D24* Pathogenic Variants Discussed in This *GeneReview*

Phenotype	DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
DOORS	c.118C>T	p.(Arg40Cys)	NM_001199107.1 NP_001186036.1
	c.119G>T	p.(Arg40Leu)	
	c.313T>C	p.(Cys105Arg)	
	c.328G>A	p.(Gly110Ser)	
	c.724C>T	p.(Arg242Cys)	
	c.999G>T	p.(Leu333Phe)	
	c.1008delT	p.(His336GlnfsTer12)	
	c.1460dup	p.(His487GlnfsTer71)	
FIME	c.439G>C	p.(Asp147His)	
	c.1544C>T	p.(Ala515Val)	
	c.751T>C	p.(Phe251Leu)	
PME	c.1079G>T	p.(Arg360Leu)	
EIEE16	c.969_970delGT	p.(Ser324ThrfsTer3)	
	c.468C>A	p.(Cys156Ter)	
	c.686T>C	p.(Phe229Ser)	
DFNA65	c.533C>T	p.(Ser178Leu)	
DFNB86	c.208G>T	p.(Asp70Tyr)	
	c.878G>C	p.(Arg293Pro)	