Title: Glycogen Storage Disease Type IV *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. *GBE1* Pathologic Allelic Variants

DNA Nucleotide Change	Protein Amino Acid Change	Clinical Phenotype	Reference Sequences
c.38insA	p.D13fs	Classic hepatic	NM_000158.3 NP_000149.3
c.143+1G>A	Disrupts splicing	Perinatal neuromuscular	
c.288delA	p.G97fs	Perinatal neuromuscular	
Genomic 900 bp del	Exon 2-7 del	Congenital neuromuscular	
c.454G>T	p.E152X	Perinatal neuromuscular	
c.430_782del	p.V144_S261del	Perinatal neuromuscular	
Genomic 25.5 kb del; 9 bp ins	p.V144_S331del	Perinatal neuromuscular	
c.671T>C	p.L224P	Non-progressive hepatic	
c.691+2T>C	Disrupts splicing	Non-progressive hepatic	
c.691+5G>C	Disrupts splicing	Perinatal neuromuscular	
c.708G>C	p.Q236H	Childhood neuromuscular	
c.728A>G	p.H243R	Perinatal neuromuscular	
c.771T>A	p.F257L	Classic hepatic	
c.783-1G>A	p.R262_S331del	Perinatal neuromuscular	
c.784C>T	p.R262C	Childhood neuromuscular	
c.895G>T	p.G299X	Perinatal neuromuscular	
Genomic 190 bp del	Exon 7 del	Congenital neuromuscular	
c.986A>C	p.Y329S	Non-progressive hepatic, APBD	
c.993_1618del	p.T332_M539del	Perinatal neuromuscular	
c.1077insT	p.T360fs	Perinatal neuromuscular	
c.1239delT	p.D413fs	Classic hepatic / perinatal neuromuscular	
c.1279G>A	p.G427R	Classic hepatic	
c.1336-1G>A	Disrupts splicing	Perinatal neuromuscular	
c.1468delC	p.L490fs	Perinatal neuromuscular	
c.1471G>C	p.A491Y	Perinatal neuromuscular	
c.1484T>C	p.M495T	Classic hepatic	

DNA Nucleotide Change	Protein Amino Acid Change	Clinical Phenotype	Reference Sequences
c.1543C>T	p.R515C	Classic hepatic	NM_000158.3 NP_000149.3
c.1544G>A	p.R515H	APBD	
c.1558delC	p.H520fs	Perinatal neuromuscular	
c.1570C>T	p.R524X	Classic hepatic / Childhood neuromuscular	
c.1571G>A	p.R524Q	Classic hepatic; Non-progressive hepatic, APBD	
c.1604A>G	p.Y535C	Classic hepatic	
c.1634A>G	p.H545R	Perinatal neuromuscular	
c.1643G>A	p.W548X	Perinatal neuromuscular	
c.1655C>T	p.P552L	Classic hepatic	
c.1774G>T	p.E592X	Perinatal neuromuscular	
c.1883A>G	p.H628R	Childhood neuromuscular	
c.1909C>T	p.R637X	Perinatal neuromuscular	
c.1999delA	p.T667fs	Classic hepatic	
Exon 16 del	p.V685_N702del	Perinatal neuromuscular	

See Quick Reference for an explanation of nomenclature. *GeneReviews* follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org).