Title: Laing Distal Myopathy *GeneReview* Table 5

Authors: Lamont P, Laing NG

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

Table 5. *MYH7* Pathogenic Variants Discussed in This *GeneReview* or Published*

DNA Nucleotide Change	Protein Amino Acid Change	*MYH7* exon	Reference Sequences
c.1322C>T [2]	p.Thr441Met	14	NM_000257.3 NP_000248.2
c.4315G>C	p.Ala1439Pro	31	
c.4442T>C	p.Leu1481Pro	32	
c.4499G>C	p.Arg1500Pro	32	
c.4522_4524delGAG	p.Glu1508del	33	
c.4622A>C	p.Gln1541Pro	33	
c.4763G>C	p.Arg1588Pro	34	
c.4772T>C	p.Leu1591Pro	34	
c.4795A>C	p.Thr1599Pro	34	
c.4807G>C	p.Ala1603Pro	34	
c.4823G>C	p.Arg1608Pro [1]	34	
c.4835T>C	p.Leu1612Pro	34	
c.4850_4852delAGA	p.Lys1617del [1]	34	
c.4906G>C	p.Leu1636Pro	34	
c.4937T>C	p.Leu1646Pro	34	
c.4985G>C	p.Arg1662Pro	35	
c.4987G>C	p.Ala1663Pro	35	
c.5005_5007delGAG	p.Glu1669del	35	
c.5117T>C	p.Leu1706Pro	35	
c.5186_5188delAGA	p.Lys1729del	36	
c.5186_5188dupAGA	p.Lys1729dup	36	
c.5377-5379del	p.Leu1793del [1]	37	
c.5401G>A	p.Glu1801Lys [1]	37	
c.5566G>A	p.Glu1856Lys [1]	38	
c.5740G>A	p.Glu1914Lys [1]	39	

* See especially Lamont et al [2014].

None of the MPD1-associated variants are present in the ExAC Browser alleles.

Note on variant classification: Variants listed in the table have been provided by the author(s). *GeneReviews* staff have not independently verified the classification of variants.

1. See Laing Distal Myopathy, Genotype-Phenotype Correlations.

References

Lamont PJ, Wallefeld W, Hilton-Jones D, Udd B, Argov Z, Barboi AC, Bonneman C,. Boycott KM, Bushby K, Connolly AM, Davies N,. Beggs AH, Cox GF, Dastgir J, DeChene ET, Gooding R, Jungbluth H, Muelas N, Palmio J, Penttila S, Schmedding E, Suominen T, Straub V, Staples C, Van den Bergh PYK, Vilchez JJ, Wagner KR, Wheeler PG, Wraige E, Laing NG. New mutations widen the phenotypic spectrum of slow skeletal/β-cardiac myosin (MYH7) distal myopathy. Hum Mutat 2014;35:868-79