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Long-term follow-up and characteristic pathological findings in severe nemaline myopathy due to LMOD3 mutations.

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Abstract

We describe the long-term follow-up of a patient with severe nemaline myopathy due to a novel homozygous mutation in the Leiomodin 3 (LMOD3) gene and describe the histopathological characteristics of the disease. The patient presented at birth with hydrops fetalis, multiple joint contractures, severe generalized muscle weakness, no movement, and respiratory insufficiency. At eight years of age, she had bilateral ophthalmoplegia, visual impairment, multiple contractures, and scoliosis, and is dependent on a home mechanical ventilator and gastrostomy. Except for slight head nodding, she has no voluntary movements. Whole-exome sequencing revealed a homozygous one-base duplication in the LMOD3 gene (c.882dupA, p.Asp295Argfs*2), which would result in a truncated protein. Muscle biopsy in the girl and an unrelated patient homozygous for LMOD3 p.Glu357* showed characteristic morphology of the nemaline rods. Many rods appeared as fragments of thickened Z-discs, frequently in pairs, which were interconnected by short thin filaments. Although not specific, this may be a morphological hallmark of LMOD3-associated nemaline myopathy.

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KEYWORDS: LMOD3; Morphological hallmark; Nemaline myopathy; Novel mutation; Severe congenital form

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