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A childhood-onset nemaline myopathy caused by novel heterozygote variants in the nebulin gene with literature review

Qi Wen [1], Xueli Chang [2], Junhong Guo [3]

Affiliations

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Abstract

Nemaline myopathy, a rare congenital myopathy, is characterized by generalized muscle weakness, hypotonia, respiratory insufficiency, and the presence of rod structures on muscle biopsy, which is caused by mutations in at least 13 known genes. A patient showing gradually deteriorated proximal muscle weakness and rod-shaped structures found in muscle fibers was suspected of having nemaline myopathy, following by the next-generation sequencing. We report two novel compound heterozygous variants in nebulin gene in a family residing in China. One is an intron event caused by an underlying variant at the + 3 position of the donor site. Another is a novel nonsense variant, which may lead to the end of protein translation and have a significant impact on protein function. The pathogenicity of this novel compound heterozygous variant remains to be verified. Variants reported here could help to diagnose NM for clinicians.

Keywords: Childhood-onset nemaline myopathy; Nebulin gene; Novel compound heterozygote.

Supplementary concepts

Nemaline Myopathy 2

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