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Format: Abstract

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Nemaline myopathies: State of the art.

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Abstract

Nemaline myopathy (NM) is one of the most common forms of congenital myopathy. The condition is defined by the histopathological finding of nemaline bodies (rods) on muscle biopsy and is associated with hypotonia and muscle weakness. The clinical spectrum encompasses lethal forms presenting in the neonatal period with profound weakness and less severe congenital diseases of later onset. NM is significantly heterogeneous from a genetic point of view, and its inheritance can be autosomal-dominant (AD), sporadic or autosomal-recessive (AR). To date, 11 genes encoding proteins of skeletal muscle thin filaments, Kelch domain-associated proteins and an unconventional myosin have been implicated in NM. The mechanisms leading to nemaline body formation and muscle weakness are still largely unclear. This report reviews the clinical, histopathological and genetic features of NM, with a focus on some of the recently discovered forms.

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KEYWORDS: Congenital myopathy; Nemaline myopathy; Skeletal muscle pathology

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