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Recent advances in nemaline myopathy

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Abstract

The nemaline myopathies constitute a large proportion of the congenital or structural myopathies. Common to all patients is muscle weakness and the presence in the muscle biopsy of nemaline rods. The causative genes are at least twelve, encoding structural or regulatory proteins of the thin filament, and the clinical picture as well as the histological appearance on muscle biopsy vary widely. Here, we suggest a renewed clinical classification to replace the original one, summarise what is known about the pathogenesis from mutations in each causative gene to the forms of nemaline myopathy described to date, and provide perspectives on pathogenetic mechanisms possibly open to therapeutic modalities.

Keywords: Clinical; Congenital myopathy; Functional; Genetics; Nemaline (rod) myopathy; Pathogenesis.

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