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Format: Abstract

Semin Pediatr Neurol. 2019 Apr;29:55-70. doi: 10.1016/j.spen.2019.01.007. Epub 2019 Feb 10.

Myopathology of Congenital Myopathies: Bridging the Old and the New.

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Abstract

Congenital myopathies (CM) are a genetically heterogeneous group of neuromuscular disorders most commonly presenting with neonatal/childhood-onset hypotonia and muscle weakness, a relatively static or slowly progressive disease course, and originally classified into subcategories based on characteristic histopathologic findings in muscle biopsies. This enduring concept of disease definition and classification based on the clinicopathologic phenotype was pioneered in the premolecular era. Advances in molecular genetics have brought into focus the increased blurring of the original seemingly "watertight" categories through broadening of the clinical phenotypes in existing genes, and continuous identification of novel genetic backgrounds. This review summarizes the histopathologic landscape of the 4 "classical" subtypes of CM-nemaline myopathies, core myopathies, centronuclear myopathies, and congenital fiber type disproportion and some of the emerging and novel genetic diseases with a CM presentation.

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