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Therapeutic Aspects in Congenital Myopathies.

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Abstract

The congenital myopathies are a genetically heterogeneous and diverse group of early-onset, nondystrophic neuromuscular disorders. While the originally reported "classical" entities within this group - Central Core Disease, Multiminicore Disease, Nemaline Myopathy, and Centronuclear Myopathy - were defined by the predominant finding on muscle biopsy, "novel" forms with multiple, subtle, and unusual histopathologic features have been described more recently, reflective of an expanding phenotypical spectrum. The main disease mechanisms concern excitation-contraction coupling, intracellular calcium homeostasis, and thin/thick filament interactions. Management to date has been mainly supportive. Therapeutic strategies currently at various stages of exploration include genetic interventions aimed at direct correction of the underlying genetic defect, enzyme replacement therapy, and pharmacologic approaches, either specifically targeting the principal effect of the underlying gene mutation, or addressing its downstream consequences more generally. Clinical trial development is accelerating but will require more robust natural history data and tailored outcome measures.

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