Congenital Muscular Dystrophy and Congenital Myopathy.

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Abstract

PURPOSE OF REVIEW: Congenital muscular dystrophies and congenital myopathies are a heterogeneous group of disorders resulting in hypotonia, muscle weakness, and dystrophic or myopathic features on muscle biopsy. This article summarizes the clinical and genetic aspects of these disorders.

RECENT FINDINGS: Historically, diagnoses of congenital muscular dystrophy and congenital myopathy have been made by clinical features and histopathology; however, recent advances in genetics have changed diagnostic practice by relying more heavily on genetic findings. This article reviews the clinical and genetic features of the most common congenital muscular dystrophies including laminin subunit alpha 2 (LAMA2)-related (merosin deficient), collagen VI-related, and α-dystroglycan-related congenital muscular dystrophies and reviews the most common congenital myopathies including nemaline rod, core, and centronuclear myopathies. With the increasing accessibility of genetic testing, the number of genes found to be associated with these disorders has increased dramatically. A wide spectrum of severity and onset (from birth to adulthood) exist across all subtypes. Progression and other features are variable depending on the subtype and severity of the specific genetic mutation.

SUMMARY: Congenital muscular dystrophy and congenital myopathy are increasingly recognized disorders. A growing appreciation for the breadth of phenotypic variability and overlap between established subtypes has challenged long-standing phenotypic and histopathologic classifications of these disorders but has driven a greater understanding of pathogenesis and opened the door to the development of novel treatments.

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