An Overview of Congenital Myopathies.

Mah JK, Joseph JT.

Abstract

PURPOSE OF REVIEW: This article uses a case-based approach to highlight the clinical features as well as recent advances in molecular genetics, muscle imaging, and pathophysiology of the congenital myopathies.

RECENT FINDINGS: Congenital myopathies refer to a heterogeneous group of genetic neuromuscular disorders characterized by early-onset muscle weakness, hypotonia, and developmental delay. Congenital myopathies are further classified into core myopathies, centronuclear myopathies, nemaline myopathies, and congenital fiber-type disproportion based on the key pathologic features found in muscle biopsies. Genotype and phenotype correlations are hampered by the diverse clinical variability of the genes responsible for congenital myopathies, ranging from a severe neonatal course with early death to mildly affected adults with late-onset disease. An increasing number of genes have been identified, which, in turn, are associated with overlapping morphologic changes in the myofibers. Precise genetic diagnosis has important implications for disease management, including family counseling; avoidance of anesthetic-related muscle injury for at-risk individuals; monitoring for potential cardiac, respiratory, or orthopedic complications; as well as for participation in clinical trials or potential genetic therapies.

SUMMARY: Collaboration with neuromuscular experts, geneticists, neuroradiologists, neuropathologists, and other specialists is needed to ensure accurate and timely diagnosis based on clinical and pathologic features. An integrated multidisciplinary model of care based on expert-guided standards will improve quality of care and optimize outcomes for patients and families with congenital myopathies.

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