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Congenital muscular dystrophies and congenital myopathies.

Iannaccone ST, Castro D.

Abstract

PURPOSE OF REVIEW: The purpose of this review is to provide information regarding the diagnosis and natural history of some very rare disorders: **congenital muscular dystrophies** and **congenital myopathies**. Patients with these conditions share characteristics such as early onset of weakness and severe hypotonia. Other organs such as the brain, eyes, and skin may be involved. Diagnosis depends largely on recognition of phenotype, muscle biopsy, and mutation analysis.

RECENT FINDINGS: More than 30 genes have been associated with these **diseases**, most of which have only been recognized in the past decade. Increasing availability of DNA analysis has been important in decreasing delay in diagnosis.

SUMMARY: Patients with **congenital muscular** dystrophy or **congenital** myopathy are at high risk of complications including restrictive lung disease, orthopedic deformities, seizures, cardiomyopathy, and malignant hyperthermia. Life expectancy varies with the severity of complications. Having an accurate and specific diagnosis allows the neurologist to carry out anticipatory guidance and appropriate monitoring. New hope exists for experimental treatments for **congenital muscular** dystrophy and **congenital** myopathy as our understanding of pathogenesis evolves.

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