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Congenital muscular dystrophies.

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Abstract

The **congenital muscular dystrophies** are a heterogeneous group of disorders in which weakness and dystrophic pattern on muscle biopsy are present at birth or during the first months of life. This chapter reviews the most common forms of **congenital muscular dystrophies**, including laminin α -2 (merosin) deficiency, Ullrich **congenital muscular** dystrophy, fukutin-related proteinopathy, rigid spine syndrome, and glycosylation disorders of α -dystroglycan. The latter group is often associated with neuronal migration defects including lissencephaly, pachygyria, cerebellar and brainstem abnormalities, and variable ocular anomalies. Typical clinical findings and underlying genetic defects are discussed to assist in the differential diagnosis and diagnostic work-up of patients with **congenital muscular dystrophies**. There are still no curative treatment options for patients with **congenital muscular dystrophies** but regular follow-up and symptomatic care by a multidisciplinary team considering the peculiarities of each disorder are important to maintain or improve patients' quality of life.

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