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Congenital myopathies and muscular dystrophies.

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Abstract

The **congenital muscular dystrophies** (CMD) and myopathies (CM) are a diverse group of diseases that share features such as early onset of symptoms (in the first year of life), genetic causes, and high risks for restrictive **lung** disease and orthopedic deformities. Understanding for disease mechanism is available and a fairly well-structured genotype-phenotype correlation for all the CMDs and CMs is now available. To best illustrate the clinical spectrum and diagnostic algorithm for these diseases, this article presents 5 cases, including Ullrich **congenital muscular dystrophy**, nemaline myopathy, centronuclear myopathy, merosin deficiency **congenital muscular dystrophy**, and core myopathy.

KEYWORDS: Central core myopathy; Centronuclear myopathy; **Congenital muscular dystrophy**; **Congenital** myopathy; Merosin deficiency **congenital muscular dystrophy**; Nemaline myopathy; Ullrich **congenital muscular dystrophy**

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