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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