Abstract

PURPOSE OF REVIEW: This article reviews adult presentations of the major congenital myopathies - central core disease, multiminicore disease, centronuclear myopathy and nemaline myopathy - with an emphasis on common genetic backgrounds, typical clinicopathological features and differential diagnosis.

RECENT FINDINGS: The congenital myopathies are a genetically heterogeneous group of conditions with characteristic histopathological features. Although essentially considered paediatric conditions, some forms - in particular those due to dominant mutations in the skeletal muscle ryanodine receptor (RYR1), the dynamin 2 (DNM2), the amphiphysin 2 (BIN1) and the Kelch repeat-and BTB/POZ domain-containing protein 13 (KBTBD13) gene - may present late into adulthood. Moreover, dominant RYR1 mutations associated with the malignant hyperthermia susceptibility trait have been recently identified as a common cause of (exertional) rhabdomyolysis presenting throughout life. In addition, improved standards of care and development of new therapies will result in an increasing number of patients with early-onset presentations transitioning to the adult neuromuscular clinic. Lastly, if nemaline rods are the predominant histopathological feature, acquired treatable conditions have to be considered in the differential diagnosis.

SUMMARY: Recently identified genotypes and phenotypes indicate a spectrum of the congenital myopathies extending into late adulthood, with important implications for clinical practice.

PMID: 27538056   DOI: 10.1097/WCO.0000000000000372

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