Emery-Dreifuss Muscular Dystrophy.

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Abstract
Emery-Dreifuss muscular dystrophy (EDMD) is a rare muscular dystrophy, but is particularly important to diagnose due to frequent life-threatening cardiac complications. EDMD classically presents with muscle weakness, early contractures, cardiac conduction abnormalities and cardiomyopathy, though the presence and severity of these manifestations vary by subtype and individual. Associated genes include EMD, LMNA, SYNE1, SYNE2, FHL1, TMEM43, SUN1, SUN2, and TTN, encoding emerin, lamin A/C, nesprin-1, nesprin-2, FHL1, LUMA, SUN1, SUN2, and titin, respectively. The OMIM database recognizes subtypes 1 through 7, which captures most but not all of the associated genes. Genetic diagnosis is essential whenever available, but traditional diagnostic tools can help steer the evaluation towards EDMD and assist with interpretation of equivocal genetic test results. Management is primarily supportive, but it is important to monitor patients closely, especially for potential cardiac complications. There is a high potential for progress in the treatment of EDMD in the coming years. This article is protected by copyright. All rights reserved.

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KEYWORDS: Cardiomyopathy; Contractures; Emerin; Emery-Dreifuss; Laminopathy; Muscular Dystrophy

PMID: 31840275 DOI: 10.1002/mus.26782
