Emery-Dreifuss muscular dystrophy: focal point nuclear envelope.

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Abstract

PURPOSE OF REVIEW: Emery-Dreifuss muscular dystrophy (EDMD) is caused by mutations in EMD encoding emerin and LMNA encoding A-type lamins, proteins of the nuclear envelope. In the past decade, there has been an extraordinary burst of research on the nuclear envelope. Discoveries resulting from this basic research have implications for better understanding the pathogenesis and developing treatments for EDMD.

RECENT FINDINGS: Recent clinical research has confirmed that EDMD is one of several overlapping skeletal muscle phenotypes that can result from mutations in EMD and LMNA with dilated cardiomyopathy as a common feature. Basic research on the nuclear envelope has provided new insights into how A-type lamins and emerin function in force transmission throughout the cell, which may be particularly important in striated muscle. Much of the recent research has focused on the heart and LMNA mutations. Prevalence and outcome studies have confirmed the relative severity of cardiac disease. Robust mouse models of EDMD caused by LMNA mutations has allowed for further insight into pathogenic mechanisms and potentially beneficial therapeutic approaches.

SUMMARY: Recent clinical and basic research on EDMD is gradually being translated to clinical practice and possibly novel therapies.

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