Cellular and Animal Models of Striated Muscle Laminopathies.

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Abstract
The lamin A/C (LMNA) gene codes for nuclear intermediate filaments constitutive of the nuclear lamina. LMNA has 12 exons and alternative splicing of exon 10 results in two major isoforms-lamins A and C. Mutations found throughout the LMNA gene cause a group of diseases collectively known as laminopathies, of which the type, diversity, penetrance and severity of phenotypes can vary from one individual to the other, even between individuals carrying the same mutation. The majority of the laminopathies affect cardiac and/or skeletal muscles. The underlying molecular mechanisms contributing to such tissue-specific phenotypes caused by mutations in a ubiquitously expressed gene are not yet well elucidated. This review will explore the different phenotypes observed in established models of striated muscle laminopathies and their respective contributions to advancing our understanding of cardiac and skeletal muscle-related laminopathies. Potential future directions for developing effective treatments for patients with lamin A/C mutation-associated cardiac and/or skeletal muscle conditions will be discussed.

KEYWORDS: DCM; EDMD; L-CMD; LGMD; animal models; cellular models; lamin A/C; striated muscle laminopathies

PMID: 30934932 DOI: 10.3390/cells8040291