LAMA2-related myopathy: Frequency among congenital and limb-girdle muscular dystrophies.

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Abstract

**INTRODUCTION:** Muscular dystrophy caused by LAMA2-gene mutations is an autosomal recessive disease typically presenting as a severe, early-onset congenital muscular dystrophy (CMD). However, milder cases with a limb-girdle type muscular dystrophy (LGMD) have been described.

**METHODS:** In this study, we assessed the frequency and phenotypic spectrum of LAMA2-related muscular dystrophy in CMD (n = 18) and LGMD2 (n = 128) cohorts identified in the last 15 years in eastern Denmark. The medical history, brain-MRI, muscle pathology, muscle laminin-α2 expression, and genetic analyses were assessed.

**RESULTS:** Molecular genetics revealed 2 pathogenic LAMA2 mutations in 5 of 18 CMD and 3 of 128 LGMD patients, corresponding to a LAMA2-mutation frequency of 28% in the CMD and 2.3% in the LGMD cohorts, respectively.

**CONCLUSIONS:** This study demonstrates a wide clinical spectrum of LAMA2-related muscular dystrophy and its prevalence in an LGMD2 cohort, which indicates that LAMA2 muscular dystrophy should be included in the LGMD2 nomenclature.

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**KEYWORDS:** LAMA2, laminin-α2; brain MRI; congenital muscular dystrophy; limb-girdle muscular dystrophy; muscle biopsy

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