Clinical and neuroimaging findings in two brothers with limb girdle muscular dystrophy due to LAMA2 mutations.

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Abstract

Recessive mutations in LAMA2 commonly cause congenital muscular dystrophy (MDC1A) and, rarely, limb girdle muscular dystrophy (LGMD). We report 2 brothers who presented in adulthood with LGMD due to novel mutations in LAMA2 identified by whole exome sequencing (WES). Muscle biopsy more than 30 years ago demonstrated dystrophic changes but was not available for immunoanalysis. Muscle MRI demonstrated involvement of peripheral muscle with internal sparing classically seen in collagen-VI related disorders. Extensive genetic testing, including COL6A1/2/3, was performed prior to WES. Subsequent skin biopsy immunoanalysis demonstrated laminin α2 partial absence. The phenotype of the patients was notable for novel central nervous system findings, namely bilateral signal changes in the globi pallidi, and presence of dilated cardiomyopathy (DCM). They also illustrate the similarity in muscle MRI in collagen VI and laminin α2-related disorders, both of which are due to mutations in genes encoding extracellular matrix proteins.

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KEYWORDS: Collagen VI; Laminin α2; Limb girdle muscular dystrophy; Muscle MRI

PMID: 27932089 DOI: 10.1016/j.nmd.2016.10.009

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