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LAMA2 Congenital Muscle Dystrophy: A Novel Pathogenic Mutation in Bulgarian Patient.

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Abstract

Congenital muscle dystrophies (CMD) are genetically and clinically heterogeneous hereditary myopathies mainly with autosomal recessive type of inheritance. The most common form worldwide is considered to be merosin-deficient muscle dystrophy type 1A, called MDC1A (due to laminin- α 2 defects as a result of *LAMA2* gene mutation), accounting for 30-40% of total cases of CMD. The exact molecular and clinical diagnoses, respectively, are a prerequisite for the most effective treatment; sometimes orphan drugs exist for some rare diseases. One of such drugs is Tarix, which was FDA approved and announced in 2016 for treatment of MDC1A. Here we present a patient diagnosed postmortem as having early-onset *LAMA2*-related muscular dystrophy as a result of mutations in *LAMA2*, identified by Sanger sequencing in his parents: a novel nonsense mutation c.4452T>A in exon 31, identified in the mother, and a known pathogenic nonsense mutation c.2901C>A in exon 21, detected in the father. The truncating nature of both nonsense mutations made the clinical presentation severe and the outcome fatal. Genetic analysis in such cases of muscle dystrophy is of utmost impact, because it makes the correct diagnosis with at least some specific options for treatment, makes the prognosis depending on the severity of mutation discovered, determines reproductive risk, and offers prophylaxis in the family by means of prenatal or preimplantation diagnostics.

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