Three novel recessive mutations in LAMA2, SYNE1, and TTN are identified in a single case with congenital muscular dystrophy.

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Abstract

Congenital muscular dystrophies (CMD) are a group of heterogeneous disorders. Here, targeted next generation sequencing of 168 CMD-associated genes was performed on collected clinic samples to identify potential mutations. A loss-of-function mutation (c.4676-4682delGCTGCAA; p.Cys1560Thrfs*33) of the LAMA2 gene in a consanguineous family was identified and confirmed by Sanger sequencing. The second recessive mutation in SYNE1 (c.2881C>T; p.Arg961Trp) was found in the SAP motif, which was predicted to be involved in chromosomal organization. The third homozygous mutation (c.32462C>T; p.Pro10821Leu) in TTN was mapped to the third PPAK motif of the encoded protein. Muscle biopsies of the proband showed large variations in muscle fiber size, necrotic and regenerating fibers and an increase in endomysial collagen tissue. To the best of our knowledge, this is the first case with CMD and mildly enlarged heart, carrying three novel recessive mutations in LAMA2, SYNE1, and TTN.

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