Congenital myopathy associated with the triadin knockout syndrome.

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Abstract

OBJECTIVE: Triadin is a component of the calcium release complex of cardiac and skeletal muscle. Our objective was to analyze the skeletal muscle phenotype of the triadin knockout syndrome.

METHODS: We performed clinical evaluation, analyzed morphologic features by light and electron microscopy, and immunolocalized triadin in skeletal muscle.

RESULTS: A 6-year-old boy with lifelong muscle weakness had a triadin knockout syndrome caused by compound heterozygous null mutations in triadin. Light microscopy of a deltoid muscle specimen shows multiple small abnormal spaces in all muscle fibers. Triadin immunoreactivity is absent from type 1 fibers and barely detectable in type 2 fibers. Electron microscopy reveals focally distributed dilation and degeneration of the lateral cisterns of the sarcoplasmic reticulum and loss of the triadin anchors from the preserved lateral cisterns.

CONCLUSIONS: Absence of triadin in humans can result in a congenital myopathy associated with profound pathologic alterations in components of the sarcoplasmic reticulum. Why only some triadin-deficient patients develop a skeletal muscle phenotype remains an unsolved question.

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