Non-lethal congenital hypotonia due to glycogen storage disease type IV

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Abstract

Glycogen storage disease type IV (GSD-IV) is an autosomal recessive genetic disorder due to a deficiency in the activity of the glycogen branching enzyme (GBE). A deficiency in GBE activity results in the accumulation of glycogen with fewer branching points and long, unbranched outer chains. The disorder results in a variable phenotype, including musculoskeletal, cardiac, neurological, and hepatic involvement, alone or in continuum, which can be identified at any stage of life. The classic form of GSD-IV is a hepatic presentation, which presents in the first 18 months of life with failure to thrive, hepatomegaly, and cirrhosis that progresses to liver failure, resulting in death by age 5 years. A severe congenital musculoskeletal phenotype with death in the neonatal period has also been described. We report an unusual case of congenital musculoskeletal presentation of GSD-IV with stable congenital hypotonia, gross motor delay, and severe fibro-fatty replacement of the musculature, but no hepatic or cardiac involvement. Molecular analysis revealed two novel missense mutations with amino acid changes in the GBE gene (Q236H and R262C), which may account for the mild phenotype.