A Case of Congenital Glycogen Storage Disease Type IV With a Novel GBE1 Mutation

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Abstract

Glycogen storage disease type IV (Andersen disease) is a rare metabolic disorder characterized by deficient glycogen branching enzyme activity resulting in abnormal, amylopectin-like glycogen deposition in multiple organs. This article reports on an infant with the congenital neuromuscular subtype of glycogen storage disease type IV who presented with polyhydramnios, hydrops fetalis, bilateral ankle contractures, biventricular cardiac dysfunction, and severe facial and extremity weakness. A muscle biopsy showed the presence of material with histochemical and ultrastructural characteristics consistent with amylopectin. Biochemical analysis demonstrated severely reduced branching enzyme activity in muscle tissue and fibroblasts. Genetic analysis demonstrated a novel deletion of exon 16 within GBE1 , the gene associated with glycogen storage disease type IV. Continued genetic characterization of glycogen storage disease type IV patients may aid in predicting clinical outcomes in these patients and may also help in identifying treatment strategies for this potentially devastating metabolic disorder.