

Amylopectinosis disease isolated to the heart with normal glycogen branching enzyme activity and gene sequence

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Abstract

Abstract: We report a 17-month-old female patient with a rare cause of cardiomyopathy secondary to accumulation of amylopectin-like material (fibrillar glycogen) isolated to the heart. Evidence of amylopectinosis isolated to cardiac myocytes in this patient was demonstrated by histology and electron microscopy. Glycogen content, glycogen branching enzyme (GBE) activity, as well as phosphofructokinase enzyme activities measured in liver, skeletal muscle, fibroblasts and ex-transplanted heart tissue were all in the normal to lower normal ranges. Normal skeletal muscle and liver tissue histology and GBE activity, normal GBE activity in skin fibroblasts, plus normal GBE gene sequence in this patient exclude the classical branching enzyme deficiency (type IV GSD). We believe that this is an as yet uncharacterized and novel phenotype of GSD associated with cardiomyopathy, in which there is an imbalance in the regulation of glycogen metabolism limited to the heart.