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Individual Treatment Trial of PIGV-Associated Mabry Syndrome with D-Mannose in a Young Child

We describe the first individual treatment trial with D-mannose in a young girl with PIGV-CDG. PIGV-CDG belongs to the GPI anchor deficiencies leading to intellectual disability, dysmorphic features, epilepsy, and, less frequently, organ malformations. A hallmark of the GPI anchor deficiencies is the elevated serum alkaline phosphatase (AP). Our patient carried the germline homozygous PIGV variant c.1022C>A, p. (Ala341Glu), the most commonly reported pathogenic variant leading to PIGV-CDG so far. We aimed to improve the impaired enzymatic function of PIGV through elevated substrate levels by giving D-mannose orally. We monitored the clinical status, developmental progress as well as serum AP levels. Our patient experienced no side effects. Standardized developmental testing showed better developmental progress during the 21-month treatment period with D-mannose than in the 12 months following the discontinuation of treatment. The D-Mannose treatment might have had a positive effect on the development of our patient with PIGV-CDG.