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Poster Abstract: Clinical

A Case of Early Infantile Pompe Disease with Atypical Manifestation

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INTRODUCTION

Pompe disease is a rare autosomal recessive lysosomal storage disease caused by deficiency of acid α-glucosidase (GAA). This deficiency results in glycogen accumulation in the lysosomes, leading to lysosomal swelling, cellular damage, and organ dysfunction. In early onset patients (the classic infantile form), this glycogen accumulation leads to death, usually before the age of 1 year. Some patients with early onset do not develop cardiomyopathy and their progression is slower (atypical infantile form). We reported a case with an atypical infantile form.

CASE REPORT

A 25-day-old female patient with consanguineous parents was referred to the Hospital de Clínicas de Porto Alegre due to significant hypotonia. She was born full-term, by cesarean section: weight 3070 g, length 49 cm, head circumference 36 cm, and APGAR score 7/9. At birth, she was hypotonic and needed oxygen. Newborn screening was normal. On physical examination, she had hypotonia and decreased deep tendon reflexes. Her echocardiogram was normal, CK was 117 IU/L, and abdominal ultrasound was normal. Electrocardiogram (ECG) showed sinus rhythm, right ventricular hypertrophy, and short QT interval. It was dosed GAA in leukocytes 0.60 nmol/h/mg protein (1.02–5.9) and GAA in fibroblasts 3.50 nmol/h/mg protein (21–139). Molecular examination showed a mutation of the GAA gene with Met502Val (c.1504 A>G) in heterozygosity. The patient started treatment with enzyme replacement therapy (ERT) at 4 months with a good response. At 3 years, she has a good development, wanders without support, but has developed hyperlordosis and hepatomegaly. She has no respiratory problems. Her echocardiogram and ECG are normal.

CONCLUSIONS

This patient with Pompe disease presented with an atypical early childhood form with a slowly progressive course, hypotonia, without cardiomyopathy. ERT resulted in a significant improvement in motor function.

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