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CLINICAL CASE

First case of a patient with late-onset Pompe disease: Cardiomyopathy remission with enzyme replacement therapy

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Abstract

Background: Pompe disease (PD) is an autosomal recessive disease caused by a deficiency in the lysosomal human enzyme α-alglucosidase. Among children (early onset), PD causes cardiomyopathy, whereas late-onset disease seems unrelated to a high rate of cardiomyopathy. Patients respond less to enzymatic replacement therapy (ERT) with α-alglucosidase.

Case report: This is the case of an 8-year-old female patient with symptom onset at 3 years of age (late onset) with recurrent respiratory infections and progressive muscular weakness. Diagnosis for Pompe disease (PD) was established due to evidence from muscle biopsy. At baseline, a right lobectomy was performed for bronchiectasis and necrosis. The patient developed pneumonia and received mechanical respiratory support (CPAP) for 4 weeks with absolute dependence on oxygen and BPAP. ERT with α-alglucosidase was given.

Conclusions: This patient's positive output and remarkable effects on cardiovascular and respiratory function suggest that ERT may reduce cardiomyopathy among late-onset PD patients.

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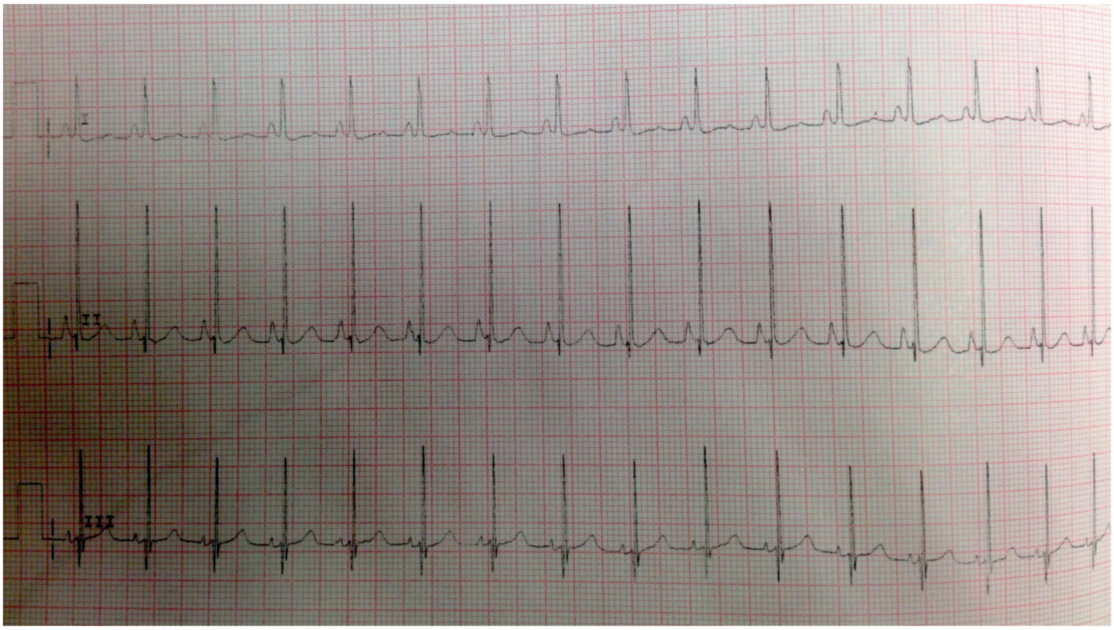
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