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Poster Abstract: Clinical

Enzymatic Replacement Therapy in Patients with Late-Onset Pompe Disease – 5-Year Follow Up

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INTRODUCTION

Late-onset Pompe disease (LOPD) is a progressive metabolic myopathy, affecting skeletal muscles, which, if untreated, leads to a serious disability and/or respiratory failure. Enzyme replacement therapy (ERT) improves muscle strength and respiratory function, and prevents disease progression. We present a 5-year follow-up of 4 patients with LOPD treated with ERT.

METHODS

Four patients with LOPD received ERT: two started treatment in 2008, the other two in 2010. All patients received recombinant human alpha-glucosidase 20 mg/kg intravenously every 2 weeks. Physical efficiency was assessed in the 6-minute walk test (6MWT), and spirometry was performed to examine FVC and FEV1. Liver enzymes, LDH, CK, and CK-MB levels were also assessed.

RESULTS

The walking distance in the 6MWT increased by an average of 9% in the first 2 years and by 21% in the first 3 years of treatment in two patients with the longest treatment compared with baseline. Similar changes were detected in spirometry: the greatest FVC increase was observed in two patients with the highest FVC values before treatment, which increased to normal values, adjusted for age and sex, with 3 years of treatment; that is, by 28% and 34%. In two other patients, FVC reached 88% and 76% of the predicted values. ERT did not affect liver or muscle enzymes levels.

CONCLUSIONS

The improvements in exercise tolerance and FVC were observed in all patients in the first 3 years of treatment, and were the greatest in patients treated the longest and with the least severe neurological and respiratory symptoms. Our research suggests that an early start of ERT results in higher improvement of respiratory and ambulatory functions.

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