

Poster Abstract: Clinical

Effects of Aerobic Exercise Training in Late-Onset Pompe Disease Before and 1 Month after Enzyme Replacement Therapy

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BACKGROUND

Pompe disease is a progressive multisystem disease caused by a lysosomal acid α -glycosidase enzyme (GAA) deficiency, resulting in lysosomal accumulation of glycogen. The late-onset form is characterized by progressive skeletal and respiratory muscle dysfunction leading to functional disability and impairment of quality of life. Enzyme replacement therapy (ERT) and treatments, such as protein-enriched diet and exercise training, have been proposed as possible countermeasures for muscle impairment in patients with late-onset Pompe disease (LOPD). However, there are no established guidelines on therapeutic exercise for individuals with LOPD, and evidence of a beneficial effect of exercise training is relatively scarce in these patients, and is mainly before receiving ERT. This retrospective study reports the results of a home-based submaximal aerobic exercise on functional capacity and pulmonary function in a patient with LOPD with mild respiratory manifestation and moderate skeletal muscle disease, prior to ERT regimen and 1 month after receiving ERT.

MATERIALS AND METHODS

The aerobic training protocol consisted of the initial guidance: 30 minutes walking on flat ground, with 60% maximum heart rate, 3–5×/week at home and bi-weekly under the supervision of a physiotherapist at our service (the Reference Center for Inborn Errors of

Metabolism [CREIM/Unifesp]). During the follow-up period, depending on the patients' tolerance, we increased the duration and frequency of walking. The patient received a booklet explaining the prescribed exercises. Before and after the training period, pulmonary function was assessed by measuring the forced vital capacity (FVC), maximal inspiratory and expiratory pressures (MIP/MEP), and peak flow (PF). Functional capacity was assessed using the 6-minute walk test.

RESULTS

The 6-minute walking distance increased (476 m before vs. 512 m after) and pulmonary function improved (FVC increased 19%, 2.6 L before vs. 3.1 L after; MIP and MEP increased 20%, 30 cm H₂O before vs. 36 cm H₂O after and 40 cm H₂O before vs. 48 cm H₂O after, respectively; PF increased 60%, 225 L/s before vs. 360 L/s after) after the aerobic training protocol during the pre-ERT period. After 1 month of ERT, the patient demonstrated an increase in walking distance (526 m) and PF (420 L/s).

CONCLUSIONS

These results suggest that aerobic exercise training has a positive effect on functional capacity and pulmonary function even before ERT, and continues to improve post-ERT. Further studies are needed to explore this form of training and to assess the long-term benefits of a home-based exercise program for patients with Pompe disease.

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