

Efficacy of telerehabilitation with games in a patient with late-onset Pompe disease

Bon M. ^{1,5}, Scapin A. ^{1,5}, Piovani P. ^{1,5}, Bizzarini E. ², Pinzini C. ³, Verriello L. ⁴, Bordugo A. ^{1,5}, Scarpa M. ^{1,5}, Sechi A. ^{1,5}

1. Regional Coordinating Center for Rare Diseases, University Hospital of Udine

2. SOC MFR - Spinal Unit, Department of Rehabilitation Medicine, IMFR Gervasutta, University Hospital of Udine

3. University of Udine - Department of Medical Area - Degree Course in Physiotherapy

4. Neurology Unit, University Hospital of Udine

5. MetabERN

INTRODUCTION

In late-onset Pompe disease, consistent physical exercise is very important to improve/maintain residual motor function, but it is often hindered by a high dropout rate due to boredom and frustration. In this study, we tested the use of Reability® Neuro, a tele-rehabilitation medical device using games, with the possibility of remote control, on a patient with late-onset Pompe disease.



CLINICAL CASE

A 40-year-old patient newly diagnosed with late-onset Pompe disease used Reability® Neuro for 6 weeks before starting enzyme replacement therapy, performing 30-minute sessions 3 times a week. Twelve rehabilitation games were selected according to the patient's clinical characteristics, divided into 4 exercises for the upper limbs, 3 for the lower limbs, and 5 for the trunk and lower limbs. Assessments were conducted at baseline t(0) and after 6 weeks of training t(1), including the 6-Minute Walking Test (6MWT) on a treadmill and the Fatigue Severity Score (FSS) questionnaire.



RESULTS

6MWT: improvement from t(0) to t(1) with an increase in self-selected speed, a significant increase in distance covered, and a decrease in average heart rate, indicating better exercise tolerance (see table).

FSS: improvement from t(0) to t(1) in all 8 items.

No increase in CPK or muscle pain was observed. High compliance and satisfaction were reported by the patient.

6MWT	t0	t1
Self-Selected Speed (m/s)	0.55	0.77
Distance Covered (m)	1980	2772
Heart Rate (bpm)	110	104

CONCLUSIONS

Although the study was conducted on a single patient, the results suggest that Reability® Neuro could be a useful tool for patients with late-onset Pompe disease to perform exercises at home using personalized game sessions under clinical observation. The advantage over traditional systems is to increase patient compliance and motivation.