

Poster Abstract: Therapeutic

Quality of Life and Participation in the Daily Life (Activities) of Adults with Pompe Disease Receiving Enzyme Replacement Therapy: 10 Years of International Follow-Up

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BACKGROUND

Pompe disease is a progressive muscle disorder for which enzyme replacement therapy (ERT) has been available since 2006. The effects of ERT have been shown on distance walked, pulmonary function, and survival. We investigated whether ERT also improves quality of life and participation in daily life in adult patients with Pompe disease.

METHODS

In an international survey, we assessed quality of life (Short Form 36, SF-36) and participation (Rotterdam Handicap Scale, RHS) annually between 2002 and 2012. Repeated measurements mixed effect models were used to describe the data over time.

RESULTS

Responses were available for 174 adult patients. In the periods before and after the start of ERT, the median follow-up times were 4 years each (range 0.5–8). The SF-36 Physical Component Summary measure (PCS) deteriorated before ERT (–0.73 score

points per year [sp/y]; CI 95% –1.07 to –0.39), while it improved significantly in the first 2 years of ERT (1.49 sp/y ; CI 0.76 to 2.21), and remained stable thereafter. The Mental Component Summary measure (MCS) remained stable before and during ERT. After declining significantly beforehand (–0.49 sp/y; CI –0.64 to–0.34), the RHS stabilized under ERT.

CONCLUSION

In adult patients with Pompe disease, ERT positively affects quality of life and participation in daily life. Compared with the progressive decline observed prior to treatment, physical health (PCS) improved and participation stabilized after starting ERT. Our results reinforce previous findings regarding the effect of ERT on muscle strength, pulmonary function, and survival.

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