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Poster Abstract: Clinical

Long-Term Interruption of Enzyme Replacement Therapy with rhGAA in Pompe Disease Leads to Irreversible Clinical Decline

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BACKGROUND

Enzyme replacement therapy (ERT) with recombinant human alglucosidase alfa (rhGAA) in Pompe disease is moderately effective and a life-long therapy is warranted. Clinical investigations of temporary ERT interruption are lacking, but might be of clinical significance (i.e. due to patient's wish, adherence issues, holidays, or problems with drug supply). In Switzerland, ERT for Pompe disease was interrupted after the federal court judged that the treatment costs for adults were greatly out of proportion. Retreatment was initiated after therapy was finally licensed.

METHODS

We retrospectively analyzed seven Pompe patients, who underwent cessation and resumption of ERT (median age 43 years). The delay from first symptoms to final diagnosis ranged from 4 to 20 years. Before ERT, all patients suffered from a limb–girdle myopathy; one was wheelchair-bound and two patients received night-time non-invasive ventilation. The demographics, clinical characteristics, assessments with the 6minute walk test (6-MWT), MRC muscle sum score, and the predicted forced vital capacity (FVC) were analyzed.

RESULTS

Initial treatment stabilized respiratory function in most patients and improved walking performance. The median duration of treatment withdrawal was 10.6 months (range 3.1–59.3 months). Afterwards, FVC declined in most, and the 6-MWT declined in all patients. Two patients needed additional non-invasive ventilatory support. Twelve months after ERT retreatment, respiratory and walking capacity improved in most patients. Aside from one patient each, none of the patients reached the levels of respiratory function and walking capacity at the time of ERT withdrawal.

CONCLUSIONS

Long-term interruption (>3 months) of ERT in Pompe disease causes a decline in clinical function. Resuming treatment only partially recovers respiratory function and walking capacity. These should be taken into account when ERT is interrupted for whatever reason.

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