

Poster Abstract: Diagnostic

Changing Characteristics of Late-Onset Pompe Disease Patients in Italy: Data from the Pompe Registry

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BACKGROUND

The Pompe Registry is a long-term, multinational, observational program designed to develop a better understanding of the natural history and outcomes of patients with Pompe disease. This analysis describes the changes in the diagnostic, clinical, and therapeutic features of Italian patients with late-onset Pompe disease (LOPD) enrolled in the Registry up to July 2010 (group 1) and to April 2013 (group 2). The patients analyzed up to 2010 are also included in the analysis up to 2013.

RESULTS

As of April 2013, a total of 116 patients from Italy were enrolled in the Pompe Registry, representing 10% of the overall Registry population. Of the 116

Italian patients, 105 (90.5%) had LOPD (symptom onset >12 months of age or ≤12 months without cardiomyopathy). Forty-nine out of 105 LOPD patients were enrolled over the time period from July 2010 to April 2013 in the Pompe Registry in Italy.

In group 1, the median ages of symptom onset and diagnosis in the Italian cohort of LOPD patients were 30 and 38 years, respectively. In group 2, the median age of symptom onset increased to 38 years, while the median age of diagnosis increased to 41 years. The average diagnostic gap (difference between age of symptom onset and age at diagnosis) has therefore decreased from 8 years to 3 years in this cohort of patients.

In group 2, the most common symptoms were proximal muscle weakness of the lower extremities (89%), trunk muscle weakness (75%), upper extremity muscle weakness (73%), and difficult ambulation (73%).

In group 2, 34% of patients required either non-invasive or invasive respiratory support compared with 25% of group 1.

In all, 68% of the group 2 patients were treated with enzyme replacement therapy with alglucosidase alfa compared with 54% in group 1. The median age of

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patients at the time of their first infusion has decreased from 53 years in group 1 to 50 years in group 2.

CONCLUSIONS

A marked increase in the number of patients with LOPD enrolled in the Pompe Registry was observed in Italy from 2010 to 2013. In Italian LOPD patients, the average diagnostic gap appears to be decreasing. A larger proportion of the patients enrolled in the Registry received treatment with alglucosidase alfa as of 2013 compared with 2010. Although the Pompe Registry is a partial collection of data on the Italian Pompe patients, these findings suggest a very active participation of Italian centers and a decrease in the diagnostic delay for patients with LOPD in Italy.

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