

Poster Abstract: Clinical

Ten Years of the International Pompe Survey: Patient-Reported Outcomes as a Reliable Tool for Studying Treated and Untreated Children and Adults with Non-Classic Pompe Disease

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Pompe disease is a rare, progressive, lysosomal storage disorder for which enzyme replacement therapy (ERT) became available in 2006. Four years earlier, the International Pompe Association (IPA)/Erasmus MC survey, an international prospective survey, was established to collect information on the natural course of the disease and its burden on patients. The survey is a collaboration between Erasmus MC University Medical Center and the IPA, and comprises an annual questionnaire that was specifically designed to assess the symptoms and problems of the disease. Here we review our results of over 10 years of follow-up, and discuss the survey's contribution to the field. Tracking 408 patients with Pompe disease between 2002 and 2013, the cumulative data reveal

the broad range of clinical manifestations that interfere with patients' lives. The survey allowed us to quantify the rate of disease progression and the positive effects of ERT on patients' quality of life, fatigue, and participation in daily life. Furthermore, it showed, for the first time, that survival is reduced in adults with Pompe disease and improved by ERT. Our results show that a patient survey can serve as a valuable and reliable tool for obtaining quantifiable information on the natural course of a rare disease, and on the effects of therapy in a large cohort. Most importantly, by working with patient-reported outcomes, the survey provides the data that are truly relevant to the patient and complementary to clinical datasets.

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