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Poster Abstract: Clinical

The Pompe Registry: 10 Years of Data

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BACKGROUND

In 2014, the Pompe Registry is recognizing and celebrating an important milestone: the 10-year anniversary of the Registry.

METHODS

Started in 2004 and sponsored by Genzyme, a Sanofi company, the Pompe Registry was established as a global, voluntary, observational database to collect and report on the natural history and outcomes of patients with Pompe disease through routine clinical practice, regardless of patient treatment status. Data currently are collected and reported by enrolling clinicians worldwide in four geographic regions: Europe, North America, Latin America, and the Asia Pacific region.

RESULTS

As of May 2014, 1292 patients in 34 countries were enrolled in the Registry. The majority (70.1%) was Caucasian, and there was an approximate equal distribution of males and females. More than three-quarters (78.9%) of enrolled patients had late-onset Pompe disease (LOPD), defined as symptom onset ≤12 months of age without cardiomyopathy or symptom onset >12 months of age; 13.8% of patients had classic infantile-onset Pompe disease (IOPD), defined as symptom onset ≤12 months with cardiomyopathy. Age at symptom onset was unknown in 7.3% of patients. In classic IOPD, the overall mean age at symptom onset was 2.9 months in 2014. The mean age at diagnosis for these patients was 4.0 months, signifying a diagnostic gap of 1.1 months. In 2014, the overall mean age of symptom onset and diagnosis for LOPD patients was 26.5 years and 33.5 years, respectively. Blood-based enzyme assays that measure acid alpha-glucosidase (GAA) activity (including dried blood spots, leukocytes, and lymphocytes), and DNA analysis were the most frequently used methods of diagnosis for patients in the Pompe Registry. Among classic IOPD patients, hypotonia was the most frequently reported symptom overall, affecting 94.7% of patients. Ambulation was impaired in 78.4% of LOPD patients in the Registry. Two-thirds (67.6%) of LOPD patients reported to have lost ambulatory capabilities, and approximately half reported using ambulation devices. Overall, 47.1% of all Registry patients had reported receiving respiratory support of some kind: 43.7% of classic IOPD patients and 48.7% of LOPD patients. LOPD patients were more likely to have non-invasive ventilatory support (41.3%) compared with patients with classic IOPD. In 2014, 82.4% of patients were reported to have ever been on enzyme

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replacement therapy (ERT) with alglucosidase alfa, 15.8% were never on ERT, and ERT status was unknown in 1.5% of patients. The overall mean age at first infusion of ERT was 7.3 months and 39.5 years in patients with classic IOPD and LOPD, respectively. The Pompe Registry has been critical in identifying new signs and symptoms not traditionally associated with Pompe disease, including arterial aneurysms, lingual weakness, oropharyngeal dysphagia, ptosis, and scoliosis.

CONCLUSIONS

With the largest enrollment of Pompe patients worldwide, the Pompe Registry continues to be a valuable resource for improving the understanding of Pompe disease in the medical community. The Registry contributes substantially to the clinical understanding of this rare disease and will continue to play a crucial role in collecting, understanding, and disseminating clinical information about Pompe disease both globally and regionally.