

Foreword

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Pompe disease, or Glycogen Storage Disease type 2, is a rare neuromuscular condition caused by deficiency of acid alpha-glucosidase (GAA) enzyme activity, transmitted in an autosomal recessive manner.

Pompe Disease can occur at any age and presents with variable clinical severity; it is a debilitating disease that can result in a considerable shortening of life expectancy. Over the past decade, the knowledge about this disease has grown exponentially, as a result of the efforts by several research groups active all over the world, either in the clinical field or in basic sciences. There have been major advances in our understanding of the characteristics of disease onset and progression, as well as the multidisciplinary complications and the optimization of treatment, which can modify the natural course of disease and improve the quality of life of patients. Enzyme replacement therapy with alglucosidase alfa (Myozyme®) has been available since 2006, and treatment has had a significant impact on the management of the disease.

The 7th European Symposium ‘Steps Forward in Pompe Disease’, took place in Turin, Italy, at the Lingotto Conference Centre on the 21st and 22nd of November 2014. The main objectives were to share and discuss important research advances and to bring together the clinical and scientific community working on and interested in the treatment of patients with Pompe disease.

Due to the active participation of 24 faculty members and 179 delegates from 38 countries from across the globe, the multifaceted aspect of the disease has been investigated and new areas for future research have been identified. Moreover, the representation by the International Pompe Association (IPA) and the Italian Glycogenoses Association (AIG) added additional value to this meeting.

The Scientific Program included four sessions: the first, ‘State of the Art in Pompe Disease’, was dedicated to the present knowledge on Pompe disease, covering infantile and late-onset presentations, respiratory and multisystemic symptoms, and a summary of eight years of experience with alglucosidase alfa treatment. The second, ‘Improving the Management of Pompe Disease’, involved the strategies to influence the course of the disease and the response to therapy, including standards of care, diet, physical activity, immunologic response, and use of MRI as a collateral diagnostic tool.

The third session was dedicated to specific challenges in the Pompe field, divided in three parallel workshops with smaller groups in order to favour interactive discussions. Workshops included ‘Differential diagnostic challenges’, dedicated to the discussion of difficult cases with complex differential diagnoses; ‘the therapeutic challenge’, which focused on the decision of when to start treatment with enzyme replacement therapy; and ‘Controversies around diagnostic approaches’, which concentrated on tools used to shorten the diagnostic delay and indications for studies on vascular malformation in patients with Pompe disease.

The fourth session, ‘Scope in the Future’ included ‘Key developments in improving the treatment of Pompe disease,’ which was entirely dedicated to the next steps with current research, covering topics from newborn screening approaches to the identification of modifying genes and gene therapy.

Two other significant sessions within the meeting deserve to be emphasized: the Keynote Lecture, ‘New pathogenetic mechanisms linking autophagy to Pompe disease’ and the ‘Late-breaking news’ session with the presentation of early, unpublished results

from three studies on the efficacy of therapies in Pompe disease.

The involvement of the participants and the increasing interest in research in the field was clearly reflected by the submission of an impressive number of scientific abstracts, with the presentation of 50 posters with authors from 18 countries. Of these, 22 were on clinical themes, 17 on diagnostic issues and 11 on therapy. The posters gave ample opportunity for

participants to share and discuss their own experiences and scientific perspectives.

The Steps Forward in Pompe Disease Symposium represents an important moment for the world medical community working on Pompe disease, and constitutes an incentive to a continuous exploration of established and new aspects of this complex disorder, always primarily aimed to improve the prognosis and quality of life of patients with Pompe disease.