

## **Chairman's report 2009**

### **1. Governance**

#### *Executive board members*

In November 2009 Ria Broekgaarden chose not to stand for re-election as Secretary to the IPA and left the IPA Executive Board. Ria has been a major power in the organization since it was founded in 1999; both in forcefully representing the needs of the patient community and, with equal diligence, in ensuring that the administration and governance of the IPA met its legal and ethical obligations. Ria will continue to have an active interest in the organization as she joins other elite members on the IPA advisory board.

We are fortunate that Mrs Wilma Treur was nominated for an Executive Board position at our AGM in Munich; Wilma not only add continuity to the representation of the Netherlands on the Board, but also has legal training and so will be a great asset to the governing body.

#### *Meetings*

Quarterly IPA board meetings were held throughout the year by teleconference. The AGM is a rare chance to meet face-to-face; except sadly for our Australian board member, Helen Walker who joins our meetings by a telephone link.

In addition we held quarterly teleconferences with both Genzyme and Amicus to discuss their treatment programs for Pompe disease and collaborative ventures.

#### *Staff*

Paula Waddell and Marsha Zimmerman have each worked part-time for the IPA in their separate roles. Paula has continued to provide excellent administrative support whilst Marsha is in the process of maintaining and updating our extensive publications.

#### *Advisory Board*

The IPA has an advisory board whose members assist with the IPA decision process when called upon by the Executive Board. Steps have been taken to formalize their relationship by the creation of specific byelaws within the IPA governing documents.

We are very pleased to welcome Ria Broekgaarden and Kevin O'Donnell onto the Advisory board in 2009. Kevin is a co-founder of the IPA and past executive board member.

#### *Scientific Advisory Board*

The IPA often consults with several world leaders working in Medical Science for Pompe disease; either in research or clinical practice or both. The IPA Board have decided to formalize the present loose arrangements to create a scientific advisory board who we may convene to scrutinise research proposals, to design IPA scientific meetings or simply to offering advice to the Executive Board and explain esoteric medical concepts in lay-terms.

#### *Membership*

The number of countries now affiliated to the IPA has grown to 39. Contact details of all these organizations or individuals can be found on our website, [www.worldpompe.org](http://www.worldpompe.org).

### **2. Myozyme/Lumizyme supply**

#### *Myozyme stakeholders working group (MSWG)*

Throughout the second half of 2008 board members of the International Pompe Association worked closely with Genzyme and expert treating physicians to monitor the global Myozyme

supply situation. Our main objective was to prepare a plan of action to modify patient demand should inventories become so tight as to jeopardise the treatment of the most vulnerable infants, children and adults within the Pompe community.

As it turned out, the disruption to supply was restricted to a single skipped infusion in early 2009 for most adults.

#### *IPA on-line survey*

The IPA was aware that there were noticeable effects from this delay in treatment and so we decided to survey a sample of the patient community to make an assessment of the true impact. Given the amount of time and effort required to prepare the Myozyme Stakeholders Working Group (MSWG) guidelines, and also the preparation of a global communication plan, we hoped to assess the quality of these through the same survey.

We received 120 individual responses representing over 10% of the global patient population receiving Myozyme infusions. A summary report is posted on the IPA website, but the main conclusions were:

- The communications plan was well designed and had the intended effect of informing the majority of patients worldwide, to prepare them for the supply interruption and to provide them with the information they needed to prior to the event.
- An unacceptable number of patients, severely affected by Pompe disease, were not exempted from the recommendations.
- It was unacceptable that patients in relatively stable condition were able to avoid missed infusions whilst others, more severely affected, missed one or more treatment.
- It was apparent that a large number of respondents were affected both psychologically and physically by the event and that it took several weeks for them to recover from its effects; some still felt damaged by the episode several months later.

#### *Lumizyme*

The continued failure of Genzyme to have the 2000l product approved by the FDA after nearly 3 years is a great concern to the IPA. There are many patients waiting to start treatment but the ATAP program is closed to newly diagnosed adults. A further setback in November 2009 means that Genzyme now has to consider other routes to approval for the Belgian (4000l) product for commercial use in the US.

### **3. Publications**

#### *Pompe Connections*

Work continues to identify gaps in the Pompe connections and to prepare new leaflets. New subjects being considered include:

- Mobility aids
- Ptosis (particularly eyelid droop)
- Drug Development Process (phases of clinical trials)
- Assisted Coughing
- Allergies and allergic reactions to ERT
- Normal Breathing and the Respiratory System
- Peripheral IV information, Central Line / Port-a-Cath information
- Scoliosis
- Swallowing
- Speech Therapy
- Tube Feeding
- Protein Drinks and Supplements

- Vaccinations and immunizations

Currently the Pompe Connections are available on-line in eight languages:

English - Dutch - French - German - Italian - Japanese - Spanish – Turkish

Work in progress includes translations into:

Czech – Croatian – Portuguese – Polish – Hungarian – Chinese – Arabic - Hebrew – Russian – Swiss - Slovenian

#### *International Genetic Alliance*

Our IPA Advisor, Maryze Schoneveld van der Linde attended a conference on “Birth Defects and Disabilities in the developing world” in New Delhi where she signed a declaration on behalf of the IPA. This declaration establishes eight principles, concerning:

- The rights of patients, families and the organisations
- Supporting the creation of Patient and Parent Organisations
- Patient and Parent Organisations role in the training healthcare professionals
- Special assistance for carers.
- Prioritizing the allocation of available resources
- Developing expert counselling and other psychological support
- The potential of those who benefit from new treatments to lead long and fulfilling lives
- A sustained programme of investment in biomedical research

The declaration was co-signed by the following organisations:

- International Genetic Alliance (IGA)
- Association of Genetic Support of Australia (AGSA)
- Brazilian Genetic Alliance (BGA)
- European genetic Alliances’ Network (EGAN)
- Genetic Interest Groups in Great Britain (GIG)
- World Alliance Neuromuscular Disorder Associations (WANDA)
- South African Inherited Disorder Association
- Indian Association of Muscular Dystrophy