



“A POSITIVE OUTLOOK”



International Pompe Association

ANNUAL REVIEW

JANUARY 2008



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### Cover Painting

#### **Artist: Sean Thomas Ackley**

**Sean:** This piece is about creating a positive outlook on anyone's situation, not only those suffering from Pompe Disease. The positive attitude I gained from Juan was the driving force of this image.

**Juan:** With hope we can transcend and rise above challenges in life, Sean's work conveys my message of hope. No one can see what tomorrow will bring, but with hope inside our hearts we can always get through today to see what tomorrow will bring.

# Review of 2007

The IPA year started in November 2006 with an excellent conference organised by the AMDA in San Antonio, Texas. Here we held our 2006 AGM where Randall House and Maryze Schoneveld van der Linde retired from the Board and we welcomed Tiffany House, Helmut Erny and Thomas Schaller as new board members.

So the board remains strong and has regular teleconferences to discuss IPA activities and to prepare for the regular confidential teleconferences that we hold with Industry. Randall and Maryze remain as highly valued advisors to the board and, indeed Maryze is invaluable with her continued work organising the translations of the Pompe Connections and also supporting patients' access to treatment in Europe, Asia and the Middle East.

Linda Paré and Juan Magdaraog agreed to work with the IPA to help with publicity and raising awareness.

One of my first duties as chairman was to revisit the IPA mission statement, Aims and objectives. Together we simplified these and created our first Annual Review document (available from the IPA website) to mark where we had got to and where we would like to go with the IPA. This is also part of a policy to be more open about the activities of the IPA in order to attract more interest in our work.

The Annual Review set forth some of our objectives for 2007 and I am pleased to say that many of our ambitions are being realised: Newborn screening is, I believe, almost at a stage where it could be implemented if national health providers have the will. The importance of multidisciplinary care is being implemented by many expert centres which we fully support. The IPA has monitored clinical trials and has issued statements to help patients understand, for example, the importance of the LOTS extension. The IPA also argued successfully for a change in the inclusion criteria for the Dose-Ranging study, a post-marketing requirement of the FDA and originally to be held in US centres only. Through our intervention poorly-responding patients in Canada and Australia were also able to experience an increase in their Myozyme dose.

The IEG (IPA, Erasmus MC, Genzyme) online questionnaire project, following on from the original paper survey by the Erasmus MC through IPA funding, is making progress; hopefully we will hear more about this in the coming months.

We have recently commissioned the redesign of our website using professional designers based in the Philippines and we hope to have the new informative, accessible and attractive site published in the first half of 2008. We have also started to investigate the use of the Internet as a means of sharing important information between national patient groups. Populating and maintaining jointly owned calendars, documents, spreadsheets and images is all possible nowadays using the internet.

We are also creating a small brochure in hardcopy to publicise the work of the IPA. The IPA believes in working closely with industry in order to closely monitor developments and best represent the views and needs of patients early in the development process. We have a long-standing relationship with Genzyme but we are also developing a close working relationship with Amicus Therapeutics who is currently researching small-molecule (oral) therapies for Lysosomal Diseases.

The IPA and Genzyme have agreed to an informal collaboration plan to help us achieve our ambitions. For example we have an up-to-date list of IPA affiliates and their corresponding Genzyme offices to help us facilitate early intervention of newly diagnosed patients. This is an important function of the IPA and something I am

particularly proud of. Several times per year we will be contacted by a distressed family member who found us through the internet. By being able to put the family in contact with both a national support group and a local Genzyme office, and helped by direct contact with the treating physician, we are able to accelerate the patient's access to therapy and also provide support for the anxious family. Indeed we often manage to put all necessary parties in contact with each other within a couple of days. By extending and improving our network of patient groups throughout the world I would hope to see this function become much more widespread.

It came as a surprise to the IPA Board this summer; to learn that in the Spring of 2006 the FDA had not approved the large-scale production method used for commercial Myozyme throughout the rest of the world. Instead it had approved a smaller scale production method that was used to produce drug for the infantile trials. A consequence of the higher than expected demand for Myozyme in the US, together with delays in getting FDA approval for the large-scale material, led to the fear of severe supply problems that could be alleviated only by transferring patients onto the Myozyme Temporary Access Programme (MTAP). The AMDA supported patients in the US admirably and the IPA supported them through joint teleconferences with Genzyme and the United Pompe Foundation (UPF).

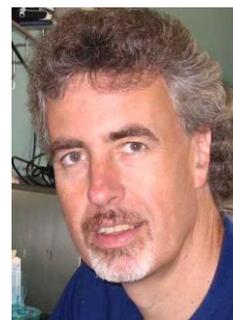
During the early stages of encouraging patients to transfer onto MTAP, an article appeared in the Wall Street Journal unfavourably comparing the larger production method with that approved by the FDA. In order to rapidly dispel the concerns of patients receiving the large-scale Myozyme and those of patients considering the transfer onto MTAP, the IPA made on-line statements, encouraged personal stories from patients outside of the USA and published an interview with Dr Ans van der Ploeg, a world expert and treating physician in Europe. Thankfully the predicted backlash from the article did not materialise.

As well as those less-welcome diversions the IPA has continued with its core work of providing information and support globally. Thanks to the persistent dedication of Maryze we have extended the number of translations of the Pompe Connections, they are now available on-line in eight languages with three in progress.

We have continued to identify patients in a large number of countries, and we have supported patients access Myozyme either through ICAP or their health providers. Whilst we cannot fund medical equipment for patients we are sometimes asked to help facilitate the transport of medical equipment across national boundaries. This year we have facilitated the procurement of equipment in Australia and have arranged the transport to Iran of Ventilators donated in the Netherlands.

The IPA continues to be an important organisation, working largely behind the scenes, but dedicated to the needs of the global Pompe community. I am very well aware, from personal experience, of the demands being placed on national patient groups, especially since the market approval of Myozyme, and I thank all those representatives who have given their time to work with, and on behalf of, this small but effective association.

Allan Muir



# IPA Mission Statement

The IPA will:

- iPA** Campaign for early diagnosis and effective, affordable and safe therapies
- iPA** Strive to provide information and support to all patients, their families and others with interests in Pompe disease

## IPA Objectives

Our mission statement gives rise to the following general objectives:

- iPA** Stimulate research into the causes, treatment and prevention of Pompe disease
- iPA** Stimulate the rapid application of research into the causes, treatment and prevention of Pompe disease
- iPA** Promote early and accurate diagnosis and screening programs for Pompe disease
- iPA** Support and encourage national organisations to obtain approval and reimbursement for therapies from government bodies and health providers
- iPA** Encourage other organisations or individuals to establish a mechanism for all patients to gain access to therapies irrespective of their personal financial status
- iPA** Encourage the formation and support the continued development of national Pompe organisations
- iPA** Establish and maintain a key position with industry, researchers and individuals with interests in Pompe disease
- iPA** Provide educational and informative publications through electronic and other media

# Objectives for 2008

## Paid Staff

The IPA has, since its conception, been largely a volunteer-based organisation where the board and its advisors carry out its day-to-day activities. The IPA Secretariat are funded by the Dutch neuromuscular charity, the VSN, to the extent of one day per week, but their dedication to our cause has often meant that they have contributed much of their own time. The workload of the IPA had increased dramatically over the last year or so due to a number of factors; the increased awareness of Pompe Disease due to the approval of Myozyme across the globe, emerging trials programs for new therapies, and the viability of newborn-screening to name but a few. We should not forget that the IPA board and advisors are also working with their national charities where demands on their time are increasing for the same reasons.

And so the IPA is now considering the feasibility of employing staff to handle some of the workload. Initially we are considering a part-time position for one staff-member, but eventually we hope to find funds to sustain a full-time post.

The job description may include the following:

- iPA** Maintain a database of national Pompe support groups around the world
- iPA** Maintain a database of national expert centres and treatment centres
- iPA** Promote the formation of support groups where none exist
- iPA** IPA Point of Contact for patients where no local support exists
- iPA** Help with the organisation of IPA events
- iPA** Coordinate and/or prepare IPA publications (including website)

The IPA would be interested to talk with any individual or organisation that could help us achieve this objective.

## Newborn Screening

On November 5, 2007, a bill was passed in Illinois, USA, to test all newborns for five fatal childhood diseases; Pompe, Krabbe, Gaucher, Niemann-Pick and Fabry diseases. This was fantastic news and we hope that other states will follow suit in the future. Elsewhere progress is slow but advances are being made both with screening technologies and with raising awareness of the need for early detection of disease in Pompe infants. The IPA will continue to advocate newborn screening and monitor its progress.

## Research

### Trials

The completion and preliminary report on the Late Onset Treatment Study for Myozyme was received with great relief that all primary endpoints were achieved. We await the published papers with interest and hope that the results will be sufficient to allow the FDA to give official approval to Myozyme from the large-scale production facility. This should finally put an end to the US supply problem and also dilute opposition to the treatment of Late-onset patients around the world.



Amicus therapeutics completed two phase I studies of their oral therapy for Pompe Disease in October 2007. These studies showed their pharmacological chaperone for Pompe Disease, AT2220, to be safely tolerated by healthy volunteers. The IPA is working with Amicus in the design of Phase II trials by presenting the patient perspective and also by facilitating dialogue between their scientists and leading Pompe clinicians.

### Patient Questionnaire

The IPA will continue to work closely in partnership with both the Erasmus Medical Centre (EMC) and Genzyme to extend the EMC Pompe Survey; this will reach out to all patients receiving therapy. The partnership, IEG (IPA, EMC, and Genzyme) was established to oversee the project. The Patient-Reported IPA Survey will be separate from the Physician-Reported Pompe Registry and will be jointly owned by the participating patient groups, academic institutions, and companies.



## Improved Communication

### Website Improvements

In order to improve our information service to anyone with an interest in Pompe disease, we are expecting our redesigned website to be online within the next few weeks (February 2008) [www.WorldPompe.org](http://www.WorldPompe.org).



### Pompe Connections



We will continue to update the Pompe Connections brochures and the Treatment Edition will be translated into additional languages

### Improved Global Reach

We hope to encourage additional members to support regions outside of their home country in order to reduce the work load on our regional group coordinators.

### Closer Links

We will strive to form closer links with Pompe groups around the world. There are countries with very large Pompe populations with whom we have very little contact.

# IPA Structure

## Board Members



Allan Muir  
IPA Chairman  
*Association for Glycogen Storage Disease (UK)*  
*United Kingdom*

Ria Broekgaarden  
IPA Secretary  
*Vereniging Spierziekten Nederland (VSN)*  
*The Netherlands*



Helmut Erny  
IPA Treasurer  
*SHG Glykogenose Deutschland e.V*  
*Germany*

Tiffany House  
*Acid Maltase Deficiency Association (AMDA)*  
*USA*



Thomas Schaller  
*SHG Glykogenose Deutschland e.V*  
*Germany*

Helen Walker  
IPA Coordinator for Australia and New Zealand  
*Australian Pompe's Association*  
*Australia*



# IPA Structure – continued

## Advisors to the Board



**Randall House**  
*Acid Maltase Deficiency Association (AMDA)*  
*USA*

**Maryze Schoneveld van der Linde**  
Coordinator: Europe, Middle East, Africa and Asia (excluding East-Asia)  
VSN  
*The Netherlands*



## Committee Members



**Marylyn House**  
IPA Coordinator for the US, Central and South America  
*Acid Maltase Deficiency Association (AMDA)*  
*USA*

**Linda Paré**  
IPA Coordinator for Canada  
*Canadian Pompe Association*  
*Canada*



**Juan Magdaraog**  
Coordinator for East Asia  
*The Philippines*

## **P.A. to IPA Secretary**

Paula Waddell  
*Vereniging Spierziekten Nederland (VSN)*  
*The Netherlands*

## **Bankers**

Rabobank  
*Soest Baarn Eemnes, Postbus 84, 3760AB Soest-Baarn, Nederland*

## **Registered Office**

Vereniging Spierziekten Nederland  
Lt. Gen. v. Heutszlaan 6, 3743 JN Baarn, Netherlands