International Pompe Association

Annual Review

January 2007

“The Hope and Freedom of Life”

CONTENTS

Cover Painting

Artist: Helen Walker, Australia

The butterfly spends the greater part of its life in a cocoon, then it escapes and flies off to start a new life.

The different types and colours of butterflies represent our Pompe people from around the world.

The lift-off signifies the hope and the freedom of life.

Retiring Chairman's Review of 2006

2006 was a fantastic year with the market approval of Myozyme by both the European Medicines Agency (EMEA) and the US Food and Drug Administration (FDA). The hard work of the IPA board members has been very helpful in speeding up the process. At the FDA, as well as at the EMEA meeting, presentations were given by IPA members Randall House, Brian White and Maryze Schoneveld van der Linde. Their contributions were very helpful in the process. "The IPA is a strong force Genzyme has to count with" was a comment made by a Genzyme representative to our Canadian associate.

The "Pompe Connections" as well as the "Flying with Pompe Disease" brochure have been composed and put on the website and distributed in print-form to patient associations.

During 2006 the board had six IPA-Genzyme telephone conferences and besides that several IPA board telephone conferences. During these conferences and meetings ongoing and future studies are discussed. My impression is that the new Genzyme team is very supportive and committed. Other issues discussed relating to Myozyme were: reimbursement, dosage, production and pricing. Insurance issues, especially in the USA, are a hot topic at this time. Because of the high price people will quickly reach the upper level of their insurance budget.

In Berlin 2006 during a Pompe conference Arnold Reuser was finally acknowledged for his work on behalf of the Pompe community; it was a great ceremony. The same happened at Duke University, North Carolina, with Y.T. Chen and Dr. P. Kishnani.

The LOTS (Late-Onset Treatment Study) trial has been discussed frequently; it is a very important clinical trial to prove the effectiveness of the drug against Pompe disease in older patients. Governments and insurance companies are realizing that there are a lot more patients than expected. Together with Genzyme the IPA must fight the reimbursement battle. The possible lengthening of the trial will depend upon the outcome of the first analysis of data in December. The IPA discussed with Genzyme higher dosage for patients not responding well to Myozyme but we were given no guarantees.

Upon request, the IPA has intervened where problems have arisen in Israel and Australia and has had positive influence in the outcomes. Board members have given presentations at several conferences about Pompe disease and the support organizations. Much attention was given by IPA and individual Pompe associations to promote awareness amongst physicians. A lot of physicians are totally unaware of the impact of the progressiveness of the disease.

Randall House

IPA Chairman 1999 to 2006

Chairman's Message

The year 2006 will be remembered by the Pompe community as the year of hope. Myozyme, an Enzyme Replacement Therapy for Pompe Disease was given market approval in both Europe and the USA for all patients: Infants, children and adults.

Market approval does not guarantee treatment however, as many of us are too well aware. Battles must still be fought with the budget holders, be they governments or health insurance companies. And even as Pompe folk begin to access their new therapy they are presented with new challenges and unanswered questions (e.g. understanding the therapy, travel to infusion centres, anxieties about infusion reactions and dosage). Many of these worries often require the support and intervention of patient organisations. As a federation of national support groups, the IPA represents their members at a global level and is able to work with governments, insurance companies, physicians and the Biotech industry to resolve many of the issues as they arise.

We must not forget that as the commercial therapy reaches across the globe there are still a large number of patients taking part in the clinical trials for Myozyme. We have recently been informed that the Late Onset Treatment Study (LOTS) is to be extended by six months to ensure the highest quality of statistically significant data; necessary unfortunately because this trial can never be repeated. It is difficult to express our gratitude to the trials' participants, but I'm sure that they know that the data they are providing will strengthen the argument for access to Myozyme for all Pompe sufferers around the world.

As the world changes for Pompe folk, so those who represent them are reorganising to both bring forward new ideas and broaden the experience of the active members. The founding member and IPA Chairman, Randall House, retired from the IPA Board at Annual General Meeting in San Antonio. He has stepped down after six years of dedicated service to the international Pompe community, although thankfully he will remain a valuable advisor to the Board. Maryze Schoneveld van der Linde has also left the board but she too remains an invaluable and very active IPA advisor. In San Antonio we welcomed Tiffany House, Thomas Schaller and Helmut Erny onto the Board so that, in many ways, the active IPA members are now more numerous than before.

The IPA executive is growing in strength and it is our wish to include additional committee members willing to assist us in our aims. Relying as we do on voluntary help from member organisations who are already supporting their own nationals, we understand the commitment required. As Pompe folk around the world look to their new futures, so we must plan for the future welfare of this small but potent organisation, the IPA.

Allan Muir

Chairman

IPA Mission Statement

The IPA will:

Campaign for early diagnosis and effective, affordable and safe therapies

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:

Stimulate research into the causes, treatment and prevention of Pompe disease

Stimulate the rapid application of research into the causes, treatment and prevention of Pompe disease

Promote early and accurate diagnosis and screening programs for Pompe disease

Support and encourage national organisations to obtain approval and reimbursement for therapies from government bodies and health providers

Encourage other organisations or individuals to establish a mechanism for all patients to gain access to therapies irrespective of their personal financial status

Encourage the formation and support the continued development of national Pompe organisations

Establish and maintain a key position with industry, researchers and individuals with interests in Pompe disease

Provide educational and informative publications through electronic and other media

Objectives for 2007

Awareness Campaign

Now that a treatment for Pompe disease exists there are a number of areas that take a higher priority than before. We want to make countries and their governments and health providers aware of the importance of these areas.

Newborn Screening
It is well documented how vital it is that Pompe infants commence therapy as soon as possible after birth to ensure the most favorable outcome. Diagnostic techniques for Pompe disease have been radically improved over the last year or two such that Newborn screening has now become a realistic prospect. The IPA will be closely monitoring progress with national Screening programs and will encourage the inclusion of tests for Pompe disease.

Multidisciplinary Care
Infants from the early clinical trials have been receiving therapy for several years; in that time it has become clear how important it is to have a multidisciplinary approach to their care. If they are to reach their full potential then these very special children must be given the very best attention in their early years. Pompe adults also need assistance from a broad range of specialties, whether receiving ERT or not and the IPA will make physicians and Pompe families aware of a paper entitled “Pompe Disease Diagnosis and Management Guidelines” which has recently been published by the American College of Medical Genetics. This timely and valuable document may be viewed at: www.acmg.net/resources/policies/Pompe_Disease.pdf.

The guidelines include recommendations in the following areas:

Diagnostics
Cardiology
Pulmonary
Gastrointestinal/Nutritional
Musculoskeletal/Functional/Rehabilitation
Neurology
General Medical Care
Surgery Anesthesia

Living with Pompe Disease DVD
This European DVD of patient testimonies will be sent out to patient groups and professionals to raise awareness of Pompe Disease and the new possibilities presented by the approved therapy.

Research

Trials
The IPA will continue to monitor closely the Late Onset Treatment Study and any other clinical trials that may be ongoing or recruiting patients. The IPA will continue to represent the best interests of the enrolled patients to the treating physicians and the trial sponsors.

Patient Questionnaire

The design and analysis of the IPA survey is reported in the PhD thesis of Marloes Hagemans, Erasmus Medical Centre, Rotterdam. The IPA will endeavor to produce a comprehensive overview of the outcome of the questionnaire for distribution to patients.

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. A new partnership, IEG (IPA, EMC, and Genzyme) has been 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 all people with interests in Pompe disease we are planning improvements to our website www.WorldPompe.org.

We plan to use a professional design service to provide a valuable, attractive and accessible resource.

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. Currently one member supports the whole of Europe and Asia. We want to divide the globe into smaller manageable regions.

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 (Chairman)
Association for Glycogen Storage Disease (UK), United Kingdom

Ria Broekgaarden (Secretary)
Vereniging Spierziekten Nederland (VSN), The Netherlands

Helmut Erny (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 (Coordinator for Australia and New Zealand)
Australian Pompe's Association, Australia

IPA members acting as advisors to the Board

Randall House
Acid Maltase Deficiency Association (AMDA), USA

Maryze Schoneveld van der Linde (Coordinator Europe and Asia, excluding E. Asia)
VSN, The Netherlands

Committee Members

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

Linda Paré (Coordinator for Canada)
Canadian Pompe Association, Canada

Juan Magdaraog (Coordinator for East Asia)
The Philippines

P.A. to IPA Secretary

Paula Waddell
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