

CHAIRMAN'S REPORT

July 2013

Allan Muir, IPA Chairman

PEOPLE

The absolute highlight of 2012 was the well-deserved decoration of Dr Arnold Reuser on August 30th. Dr Reuser was decorated as a Knight in the Order of the Dutch Lion by decree of Queen Beatrix of the Netherlands. The award was given for his outstanding achievements and for pioneering the first drug for an inherited muscle disease (Pompe).

A desperately sad day for the Pompe community came just one month after the decoration of Dr Reuser when we learned of the passing of Helen Walker, President of the Australian Pompe Association and Board Member of the IPA. Helen passed away at the age of 72 and had been member of our board for over 10 years.

Helen made sure that her Australian compatriots and her friends from New Zealand were never forgotten. She fought constantly in the interest of Pompe patients in her country and New Zealand, and also of those in other countries; especially around South East Asia. She was an active contributor in the writing of the Pompe Connections and the Pompe Connections Treatment Edition to help educate patients about their disease. We will greatly miss her, especially her passionate contributions to our deliberations.

PATIENT GROUPS

We are constantly encouraged by the work of national Pompe patient organisations; below are just a few examples from around the world.

We are especially heartened to see a group of Chinese Pompe patients and families come together to form a Pompe Rare Disease Care Centre in March this year.

Two Balkan patient groups from Croatia and Serbia joined together to share experiences, and ideas for raising awareness and improve their networking.

On June 1st the Indian LSD Support Society held their second Pompe Awareness day achieving significant coverage in newspapers and on TV.

The Australian Pompe Association and individuals in Australia and New Zealand have attracted a great deal of local and national media attention in their fight to access reimbursement for Myozyme.

INTERNATIONAL POMPE DAY

Given the clear desire of the international Pompe community to recognise a day in year to raise public awareness, the IPA is coordinating an International Pompe Day for 2014. The date for this will be the date of Dr J C Pompe's death, April 15th. Although it will be our official day for promoting awareness, individual countries will be free to use their own established dates or other days near to April 15th to avoid conflict with other national events.

NEWBORN SCREENING IN THE USA

On May 17th 2013 the Federal Advisory Committee voted to recommend Pompe Disease to the Required Uniform Newborn Screening Panel (RUSP). This means that they are recommending that it be mandatory for all states to screen for Pompe Disease.

This is a huge step forward in making newborn screening for Pompe a reality in the US, and was made possible through the support of the Pompe community within the US and also from supporting representations from outside the US.

GLOBAL ACCESS TO TREATMENT

In the past twelve months we have witnessed new threats to the reimbursement of Myozyme as health authorities take a fresh look at their budgets in these times of austerity. Thankfully countries such as Poland have agreed to continue reimbursement. What is apparent from these events is that in many countries there will be new guidelines or Standard Operating Procedures introduced to govern the starting and stopping criteria for such expensive Enzyme Replacement Therapies.

Continued research at the Erasmus Medical Centre (EMC) in the Netherlands continues to make very supportive contributions to the approval process: the therapeutic effectiveness of Lumizyme/Myozyme has been proven for babies, children and adults. New assessment scales have been developed such as the Rasch-built Pompe specific activity (R-Pact) scale and we have seen many new publications and a very interesting thesis from Nadine van der Beek concerning the clinical features, disease course, and effects of enzyme therapy in people with Pompe disease.

The IPA is greatly disappointed that, despite the great efforts of the APA and new supportive and convincing evidence, the Australian authorities have yet again turned down the inclusion of Myozyme into their Life-Saving Drugs Programme.

DRUG DEVELOPMENT

Five companies are now known to be developing competing therapies for Pompe Disease, each based on Enzyme Replacement Therapy (ERT). The IPA board are in regular contact with these companies to offer our experience of previous drug trials and to present the patient perspective during the study design.

All of the new therapies offer the potential to reduce the amount of protein that needs to be infused, which translates to patient and medical benefits of better efficacy, shorter infusion times, reduced immunogenicity, and greater convenience.

GENZYME

Genzyme have at last announced that phase II trials for their next generation ERT, Neo-GAA, will commence late in 2013.

BIOMARIN

After being convinced that their pipeline product BMN 701 (a fusion of IGF2- and GAA) was shown to be superior to Myozyme/Lumizyme in their Phase I/II study, BioMarin are now planning a phase III trial that they hope will provide the evidence for FDA and EMA approvals by 2016.

AMICUS THERAPEUTICS

After their success with the oral formulation of AT2220 co-administered with Myozyme/Lumizyme, Amicus is now developing an intravenous formulation of AT2220 (AT2220-IV) co-administered with ERT in Phase 2 for individuals with Pompe disease.

In addition, an investigational next-generation ERT (AT2220 co-formulated with a proprietary rhGAA enzyme) is in preclinical studies to improve the properties of the rhGAA enzyme itself while incorporating AT2220 as a small molecule stabilizer.

OXYRANE

Oxyrane is developing an ERT for Pompe through a yeast expression host, *Yarrowia lipolytica*. This is claimed to improve protein expression and to consistently produce proteins with desirable sugar structures (glycans). Clinical studies are expected to start before the end of 2013.

CALLIDUS BIOPHARMA

A new venture on the scene is Callidus, founded by Dr. Hung Do who brings experience from both Genzyme and Amicus. Callidus' technology platform is based on attaching a form of the naturally occurring Insulin-like Growth Factor (IGF-2) to enhance the targeting of therapeutic enzymes to the relevant cells in the body.

POMPE CONNECTIONS

The IPA continues to work closely with our international affiliated patient groups to update and translate the Pompe Connection brochures.

The updated English brochures have been translated into nine languages: German, Spanish, Italian, Dutch, Russian, Greek, Chinese (in progress; Traditional and Simplified), Korean and Arabic. Translators are still required, however, for Turkish, Portuguese, Polish and Hebrew.

As new brochures and translations are completed they are made available for download through our website at:

www.worldpompe.org/pompe-disease/publications

INTERNATIONAL GSD CONFERENCE, HEIDELBERG

<http://conference2013.worldgsd.org>

In November 2013 The IPA is supporting the German patient organisation for Glycogen Storage Disease to host the first international GSD conference for scientists and patient representatives. IPA board-member Thomas Schaller is the driving force for this event and so we are sure that it will be a great success and will bring together GSD patient groups from around the globe.

The IPA encourages all member organisations to contribute to this conference with their ideas and experiences.

The IPA will hold its Annual General Meeting (AGM) during this conference and all groups affiliated to the IPA are strongly encouraged to attend.