

ISMRD – the International Advocate for Glycoprotein Storage Diseases

Annual report to Board of Directors for the 2008 year – plus commentary to the date of our meeting 12 May 2009.

The most significant events during 2008 and in the following months up to May 2009, are a mixture of positive and negative.

- The financial and economic situation had a direct impact on our fundraising efforts and we were unable to secure sufficient funds to continue the role of executive director.
- Loss of the involvement of several board members led to continuing problems organising efficient administration for ISMRD, as well as problems with the operation of committees.

Despite these organizational problems we have made some good progress in our research agenda:

- A very successful consensus development meeting was held in November 2008 to work on protocols for use of Bisphosphonate therapy in Mucopolidosis. A grant of NZ\$40,000 was received from the UK MPS Society to assist with this consensus development meeting.
- A grant of NZ\$60,000 was obtained by Jenny Noble from the AMP foundation to do work related to the consensus meeting, and to help fund the inclusion of Australian and New Zealand families in the natural history study.
- The relationship with the Greenwood Genetic Clinic has been maintained. Progress, though slow, is continuing with the natural history study. This includes support of US\$20,000 from the National MPS society directly to the Greenwood Clinic for this study, plus collaboration agreed to by Michael Beck and John Hopwood to extend this study to more countries.
- Connections we had established with researchers lead to a grant of US\$60,000 from the MPS Society to Richard and Jennifer Steet for work on a Zebrafish model of Mucopolidosis.
- A major grant application for US\$250,000 has been submitted to the NIH via the Lysosomal Diseases Network to seek funds for the natural history study. A decision is expected soon.
- Extensive background research has been done by John on newborn screening decision criteria in preparation for our advocacy aimed at getting Lysosomal diseases included in standard test panels. We are greatly assisted in this area through Barbara's role in newborn screening in the US, and our connections with the screening program in New Zealand. Close connection is maintained with the Genetic Alliance who have a researcher with particular responsibility for policy on this topic.
- ISMRD has maintained good links with a range of Lysosomal research groups and networks.
- Good connections have been established with ICORD, the international conference on rare diseases and orphan drugs. This group includes many regulators, industry, academics and officials from the US and EU, and its agenda includes a range of topics of close interest to us, such as orphan drugs regulations, research incentives, clinical trial requirements, etc.

The success we have had in making things happen since the start of ISMRD exactly 10 years ago should not be underestimated. In addition to items listed above, we have achieved significant support from the National Institutes of Health to hold two major scientific/family conferences in 2004 and 2007, with the combined value of NIH contribution for these two meetings likely to be well over US\$150,000.

However, most of the funds raised towards research and treatments for our diseases, do not pass through our books, and do not provide us with any overhead component. We are more likely to be a catalyst for funds to go from funders to researchers, rather than have funds come directly to us. Securing administration funds for ISMRD has been a major difficulty for us. It is a significant fact that many donors do not wish to contribute to administration overheads, but another reality is that our ability to continue to advocate for our diseases and act as a catalyst for these research funds to flow, is severely compromised by our difficulties in getting

enough money to secure our administration and advocacy functions. That was the major dilemma during the time we employed an Executive Director. Her work assisted this catalyst function well, but was unable to generate the essential administration funds to support her own role and our office functions.

A number of ISMRD families have run fundraisers on their own initiative to supplement ISMRD's own fundraising efforts, or made direct contributions to ISMRD. Over the past three years this has produced close to US\$40,000 on top of the US\$74,000 generated through the Walk/run events organised by Terri. All of these fundraising efforts have provided vital funds for us, and we are very grateful for them, but we have not been able to achieve these at a level sufficient to give us the organizational capacity we would like to have.

We must now revert to an all-volunteer operation and our busy lives and the economic problems make that more of a challenge than it was when we started 10 years ago. The key challenges for us continue to be: How to ensure efficient organisation of our accounts, receive mail, file returns, and maintain our database, fundraising activities, and our communications with families and other interested parties, so that our mission can be achieved.

John Forman
President
ISMRD