

ISMRD – the International Advocate for Glycoprotein Storage Diseases

Annual report to Board of Directors for the 2009 year

2009 was a very challenging year for ISMRD and I want to acknowledge the many hours of work the board has put into restructuring ISMRD's operations. It has been quite a challenge to organise some items across borders, and I'm very pleased with the way the board members have ensured continuity of all our administrative processes, while still managing to look towards future projects.

In this our first Pathways for 2010 you will see reports on some of the projects we have been working on and stories from families who have been raising funds to support the mission of ISMRD.

There have been some fantastic fundraisers organised throughout 2009 and the early part of 2010. The Charity Dinner held in California by the Gates family raised \$32,000US. This fundraiser helped stabilise our very precarious financial situation. Other fundraisers held were the Rock for Dakotah, Ethan's Slide Glide and Ride bash and Pam Tobey's music CD sales. These are exciting innovative ways of helping us raise funds. Thank you to all those families who have been involved. We also have financial support that comes to us via other means such as the Kimmel family monthly donations, our accounting fees covered by Mark Stark, LDNZ who are covering postage and printing, and NZORD who host the website and cafe. I also want to acknowledge all those families and friends who have sent in donations to ISMRD via PayPal, JustGive or have sent in cheques. Total including to the Gates family's big fundraiser was \$55,600US during the 2009 year. Without your valuable support ISMRD would struggle move our mission forward.

The board has recently submitted a number of grant applications for various costs and projects and we plan to continue these applications on a regular basis from now on. We hope that with persistence ISMRD will become successful in gaining grants to support the wonderful efforts of our families in raising funds for us.

Our projects during 2009 saw ISMRD supporting ML families to attend the Greenwood Genetic Centre for the second stage of the Natural History Study for ML patients, and in November 2009 Dr Sara Cathey and Dr Lucia Horowitz travelled to Australia and New Zealand to gather samples and Natural Histories from 15 affected family members. We are very appreciative of the US\$7000 grant provided in 2009 by the US MPS Society to support this work, with the rest of the costs coming from the research scholarship won by Jenny Noble in the previous year.

Our last annual report discussed a major grant application to the NIH to support Dr Sara Cathey's work on the natural history study and we are pleased to note that a grant of US\$250,000 was given via the Lysosomal Diseases network. This will support the study over the next 5 years.

Our Website and Penguin cafe have had some makeovers and upgrades, and in March this year they were transferred to a new content management system and hosting arrangements that offer more security and ease of updating. Our next plan for the website is to arrange translation into several languages. This is an important part of the International aspect of ISMRD. We have also printed new brochures and a banner for ISMRD.

We have been updating our database of ISMRD family contacts and have moved it to a system that is easier to manage. During this update we saw our database grow from 150 to 175 families. We still need to follow up on families who have moved and not updated their details. If this applies to you could you send your new contact details to Jenny Noble or use our info@ismrd.org address. That email address is now the primary contact point for ISMRD.

We continue to work on the issues that are important to our group of diseases. Lobbying on issues related to newborn screening for Lysosomal diseases has produced a commitment from New Zealand's screening advisory committee to review screening criteria. A number of US states have begun screening for a small number of other Lysosomal diseases. Both these developments have indirect benefits for us by keeping up the momentum and debate about new conditions to screen for, and we continue to keep a close eye on these developments, working towards the day when our conditions are also screened for in new born screening programs.

Our networks with other Lysosomal research groups continue to be strong. Late last year ISMRD became a partner with the EveryLife foundation set up by Dr Emil Kakkis, to look at how the research and fast tracking of orphan drugs

can be improved for rare and neglected diseases. We are also partners in the Global Genes project and you can read more about this in this edition of Pathways.

2010 is shaping up to be a very busy year for ISMRD. We are taking the opportunity to host an ISMRD family meeting in the USA immediately following the WORLD Lysosomal research meeting, in Las Vegas, on 19 and 20 April 2011. In addition to this, we are making plans for a scientific and family meeting in Europe around the middle of 2011. This will be an important opportunity to reach out to other Glycoprotein storage disease families beyond the mainly English speaking membership we currently have.

Best wishes
John Forman
President