Pathways

The Newsletter for Glycoprotein Storage Diseases

ISMRD, a 501 © not-for-profit organization, FEIN 53-2164838 | website www.ismrd.org

Our Mission and Vision

ISMRD is the leading advocate for families world-wide affected by Glycoprotein Storage Diseases.

Through partnerships built with medicine, science and industry we seek to detect and cure these diseases, and to provide a global network of support and information.

We seek a future in which children with Glycoprotein Storage Disease can be detected early, treated effectively and go on to live long, healthy and productive lives.

ISMRD supports the following disorders

Alpha Mannosidosis, Aspartylglucosaminuria, Beta Mannosidosis, Fucosidosis, Galactosialidosis, Mucolipidosis II alpha/beta (I-Cell Disease), Mucolipidosis III alpha/beta (Pseudo-Hurler Polydystrophy), Mucolipidosis III Gamma, Schindler Disease and Sialidosis

https://www.facebook.com/groups/82945687520/

https://twitter.com/ISMRD

Donations

ISMRD is a 501(c) charitable organisation based in the United States serving a global constituency. We provide our services, which include our newsletter, website, outreach activities and support of research, without requesting monthly dues or any other financial restrictions. We gratefully accept donations that will enable us to continue toward our goal of a future free of the tragic consequences of Glycoprotein Storage Diseases.

Donations can be made via our website using

PayPal Secure Payments

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From the President's Desk

By Jackie James
President, ISMRD

Christmas and Holiday greetings to the entire ISMRD family! It's difficult to believe that 2016 is almost at an end and that 2017 is just around the corner. 2017 is shaping up to be a very exciting, progressive year for ISMRD!

It is with great pleasure that we announce our very first European scientific and family conference in Rome, Italy! The conference will be held in November 2017, so get your passports ready!

Members of our Scientific Advisory Team are working hard on the final deliberations of the Mucolipidosis Research candidates. We look forward to making the announcement of the research project(s) within the next few weeks. I would like to thank everyone that worked so hard on raising funds and making this research initiative possible. The fact that we raised $150,000 is amazing!

We do continue to seek funding for further research, and for our upcoming 2017 conference, and ask that if you are able, to remember ISMRD in your giving, or fundraising efforts in the upcoming months.

We would like to extend our thanks to our board member John Forman who attended a landmark event for rare diseases at the United Nations in New York last month. John has written a summary of his visit and the significance of the newly formed United Nations Committee for Rare Diseases. This is a tremendous boost for support for us and other families dealing with rare disease.

February will be here before we know it, and we are readying for Rare Disease Day which is always held the last day of February. Check out the paragraph detailing how you can participate in the 2017 Rare Disease Day, and keep an eye out on our social media and website for ways that ISMRD will be participating.

There is also exciting news from the National Organization for Rare Diseases that the 21st Century Cures Act has been passed by the US Senate! Check out the paragraph that details what this means for families with rare disease in the U.S. We also list interesting news for France, Australia and China, as these countries work on progressing their work to help those with rare disease.

I also pause to remember those we have lost and those that have had to deal with illness or surgery. Our prayers and thoughts go out to the family of Liam Urbaghs, age 4, from the Netherlands who passed away on December 7th from ML II. We are so sorry to hear of your loss.

We also continue to keep Hunter, Zachie, Megan and Malynda in our thoughts and prayers as they recover from illness and surgery.

I want to wish each of you a Christmas season full of peace, joy and happiness, and a very happy New year. I hope to see many of you in Rome next year!

Jackie James
ISMRD Board President
Mom to Anna, ML III

ISMRD conference in Rome, Italy - 2017!

ISMRD is proud to announce the 5th international conference on Glycoproteinoses to be held in Rome in November 2017

This is ISMRD's first conference to be held in Europe! It will be held at the A. Roma Lifestyle Hotel in Rome, Friday 2- Sunday, 5 November 2017. Go to http://www.hotel-aroma.com/ for details and photos of this beautiful hotel.

The conference program will feature keynote presentations by international speakers with a strong interaction between scientific and family meetings. As usual, there will be a Welcome Reception and a Gala Dinner.

The conference will be Continuing Medical Education accredited, another first fir ISMRD. Alessandra d'Azzo will be our Scientific Chair. The Scientific Committee will comprise Dag Malm, Thomas Braulke, Marc Patterson, Stuart Kornfeld and Maurizo Scarpa

More information will be available on the ISMRD website in early 2017

ISMRD's Mucolipidosis Research Initiative

The Wagner Foundation

Irish MPS Society

Update

We have almost completed evaluation of the research proposals submitted for our $150,000 ML research fund. Our expert reviewers' comments have been received and there are just a few details to be sorted before we announce the successful bidders.

Thank you for the generous support across all of ISMRD's family network, and thanks also to our partner organizations for making this important step possible.

We expect to announce the decision before the holiday period is upon us.

Watch out for breaking news!!!!

UN Committee for Rare Diseases launched, ISMRD represented at the event

A significant event for rare diseases took place at the United Nations in New York in November this year, with the inauguration of the UN Committee for Rare Diseases.

John Forman, who is ISMRD's Vice-President for Research, was present in his role as immediate Past-President of ICORD, the International Conference on Rare Diseases and Orphan Drugs.

"I have several roles in the rare disease community", said John, "and whenever I speak at any event or forum related to rare diseases, I am very conscious that just about everything I know about rare diseases, started with the grounded experience of being a parent of twins with a lysosomal disease.

That experience, plus the many contacts I have with families in Lysosomal Diseases New Zealand, and in ISMRD, are what makes it possible for me to engage with the policy makers, health officials, and medical/science leaders."

Ramaiah Muthyala, Indian Organisation for Rare Diseases; **Megan Fookes**, Rare Voices Australia; **Rachel Yang**, Chinese Organisation for Rare Diseases; **Durhane Wong-Rieger**, Canadian Organisation for Rare Diseases; **John Forman**, LDNZ, ISMRD and ICORD at the UN Headquarters in New York

John with his wife Judith and twins Tim and Hollie, who have Alpha-Mannosidosis

The objective of the Committee for Rare Diseases will be to serve as an advocacy platform for rare diseases, increasing awareness and understanding of rare diseases within the UN and worldwide as well as stimulating collaboration among different stakeholders, including the international NGO community, major UN agencies, national governments, the academic and scientific world and the private sector.

Commentary on the meeting and its background, along with video clips of the event, notes from John's talk to the meeting and a summary that John has prepared about the meeting and its significance, can be found on the website of ICORD at this link.

What's happening around the world?

2017 Rare Disease Day

RESEARCH

28 February 2017 is the tenth Rare Disease Day. This year's theme is Research. Research is key. It brings hope to the millions of people living with a rare disease across the world and their families. The day will see thousands of people from all over the world come together to advocate for more research on rare diseases.

There are some great ideas on how you can participate on the **Rare Disease Day** website. For instance, you can tell your rare disease story at **Tell Your Story**.

USA 21st Century Cures Act approved

The 21st Century Cures Act has been passed by the US Senate. According to the US National Organisation for Rare Diseases, the Act is a "landmark medical innovation package that has the potential to help nearly every American family, including the 1 in 10 Americans with rare diseases who desperately need treatments and cures."

The bill includes many provisions that will improve the discovery, development, and delivery of orphan therapies for rare disease patients, including:

- An extension of the Rare Pediatric Disease Priority Review Voucher Program, which incentivizes the development of new therapies to help the more than 15 million children with rare diseases;
- Streamlining of US Food and Drug Administration (FDA) review of genetically targeted and protein variant therapies for rare diseases;
- Creation of funds in the amount of $4.8 billion over 10 years for the National Institutes of Health to fund the Precision Medicine Initiative, BRAIN Initiative, and the Cancer Moonshot; and
- Further expansion of the Patient-Focused Drug Development Initiative and requirements for the FDA to report on how patient experience data was used in regulatory review.

France prepares Third National Plan for Rare Diseases

France has announced its decision to launch a third National Plan for Rare Diseases.

Representatives from patient organisations, the healthcare sector, research organisations, national insurance body, industry and information, as well as the respective ministries representatives discussed the main themes to be further developed by ad-hoc working groups. A steering committee will be constituted to ensure the overall coherence and integration of the recommendations made by working groups.

With this Third Plan, France aims to build on the lessons learned from the previous ones, to consolidate their achievements, but also to go further towards a better integration between health and research, between healthcare and social care, and with European and international policies and initiatives, in order to promote equity to access to diagnosis, global care and innovation for patients.

Read more

Fast-track approval of medicines in Australia

The Australian Federal Government is changing the way new medicines are approved to ensure Australians get access to breakthrough drugs more quickly.

At the moment, Australian patients are being forced to wait up to two years longer to access some drugs than patients in the United States and Europe.

Under the changes, any drug that has been listed by a comparable overseas regulator, including the US Food and Drug Administration and the European Medicines Agency, can now be fast-tracked for approval and sale in Australia.

The Therapeutic Goods Administration (TGA) will be able to share information with those overseas regulators, meaning it would no longer have to start the lengthy approvals process from scratch, in every case.

The regulatory changes would also allow pharmaceutical companies to list their drugs in Australia at the same time as they list them in larger, overseas markets, she said.

The Government allocated $20.4 million in the May budget to implement the changes, which will be rolled out over the next two years.

Read more

Rare diseases in China

With a population of 1.4 billion, China shares the largest burden of rare genetic diseases worldwide. Current estimates suggest that there are over ten million individuals afflicted with chromosome disease syndromes and well over one million individuals with monogenic disease.

Authors of an article published in *Intractable Rare Disease Research* discuss the magnitude of the challenges the Chinese are faced with to manage their burden of rare diseases. According to the authors, despite the magnitude there is much promise due to the social support available to patients and the push towards educating government bodies and clinicians. The authors believe that with the advent of next generation sequencing, especially non-invasive prenatal testing, there is a possibility of reducing the number of individuals with rare genetic diseases over time.

The key according to them is to develop cost-effective treatments backed by government subsidies.

Read more

Christmas Giving

As the festive season approaches, and you are doing your Christmas shopping, please consider making a cash donation to ISMRD. We are the only organisation that advocates for the Glycoprotein Storage Disorders, we are a not-for-profit body, our board members are not paid and we do not charge membership fees. We work hard to progress the interests of our families, and for that we need finances. Every flyer, poster, conference, area of research costs money and you can help by making a small donation in this time of gift-giving.

Please make a donation to ISMRD now using PayPal at **Christmas Giving**

ISMRD mourns the passing of

Liam Urbaghs

Liam was four years old when he passed away

on 7 December 2016

He lived in the Netherlands, and had Mucolipidosis II

We send our deepest sympathy to his family and friends

If you know of anyone who has recently been ill or had surgery or is about to have surgery, please tell us at info@ismrd.org

Some of our Penguin children and young adults have recently been in hospital, had surgery or are awaiting surgery. Your Penguin family are thinking of you and praying for a good outcome

- Zach Haggett who has ML ll/lll and has had chest problems
- Megan Rust who has ML ll and who dislocated her hip for the second time
- Malynda Norstadt, who has ML lll and who spent several weeks in hospital, first with an infection, then with heart problems that were uncovered

ISMRD'S Sunshine Care Committee

ISMRD has a group of parent volunteers called the **"Sunshine Committee"**. Our purpose is to coordinate support for families in need. The type of support varies on the circumstance -- from birthday and weddings, an illness or death in the family, or a family experiencing surgery or a medical crisis. In any case, we provide a little "sunshine" for the family by providing flowers, encouraging messages via email, cards or a phone call -- whatever we think the family would find most helpful. In order to help others, our group relies on the support of all families because, in essence, we are all part of the ISMRD "Sunshine Committee".

If you are in need of assistance or know someone in our Penguin community who is in need, **please contact Susan Kester**. She will coordinate with the appropriate parties to determine how we can best help.

ISMRD gratefully acknowledges the following people for their very generous donations

Without this kind of support we would not be able to carry out our mission and vision for ISMRD

Ashley Pauls
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David Dollins
Johnny Chooey
Mariela Guzman
Mark Stark
Martin Woolley
Mary Kimmet
Queanbeyan High School
Rock4Dakota
Shirley Jamil
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