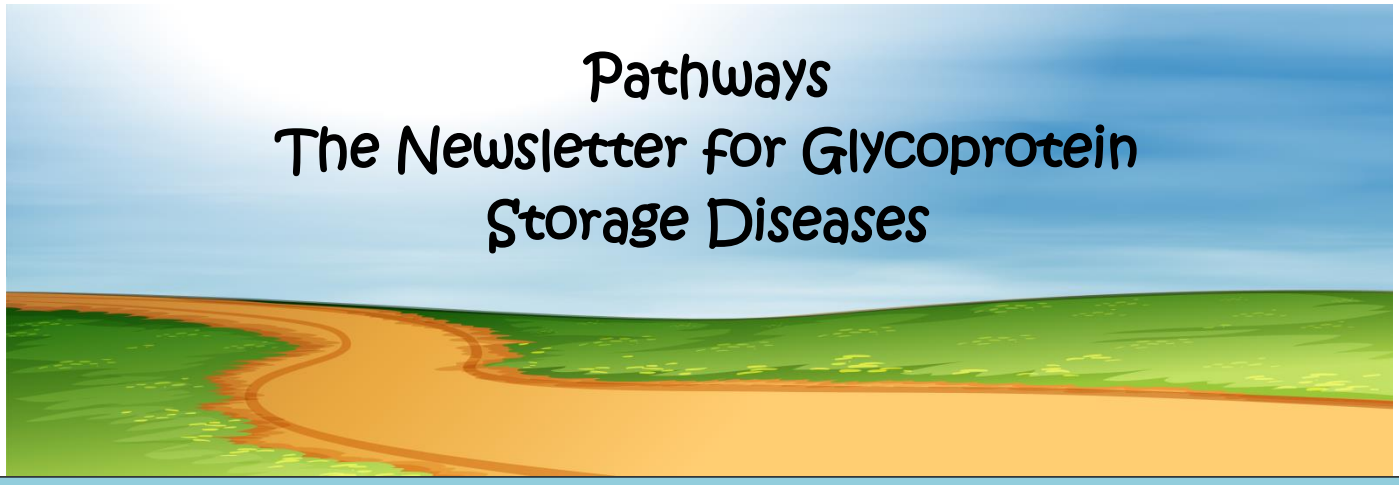

ISMRD, a 501 © not-for-profit organization, FEIN 53-2164838 | website www.ismrd.org

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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Contact Us

The International Advocate for Glycoprotein Storage Diseases

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From the President's Desk

By Jackie James
President, ISMRD

Having a child with an ultra rare disease is often isolating, and it easy to believe what is so often the case, that our rare diseases are forgotten and too small to garner the attention of researchers and scientists. For me, the past couple of years have been exciting, as I have seen several of our covered disorders catching the interest of the medical community. There is now ongoing research into enzyme replacement therapies for Alpha Mannosidosis and now there are exciting new developments for the possibility of ERT for Fucosidosis. Our editor, Carolyn Paisley-Dew, includes a very encouraging article on the new mouse model for Fucosidosis and the possible developments this can open up for an enzyme replacement therapy.

ISMRD member Shirley Jamil (mom to Sam ML III) has been working overtime on new Mucolipidosis wrist bands to raise funding for ISMRD. Shirley has written a fantastic article on how these wrist bands have travelled the world! Thank you Shirley for your hard work and enthusiasm!

It is with great excitement that we have joined forces with the MPS society in adding all nine of the ISMRD glycoprotein storage diseases to the ConnectMPS registry. Our article covers what this means and the amazing potential this offers in bringing researchers and families together.

Our "What's happening around the world" feature covers the state of rare disease management in Southeast Asia, newborn screening for Krabbe's Disease and a brief summary of a research publication on the impact of health-related quality of life of parents of children with a metabolic disease.

For any budding photographer interested in winning an iPad and other great prizes, Eurordis is offering a great photo contest! Details of how to enter the contest are listed in this newsletter. We also give the details of a wonderful project called "My *Beautiful* CHILD". This is an online project open to families where you can submit your photos and story. What a great way to highlight our rare diseases!

Until our next edition, I would like to take the opportunity to wish you and your family very best wishes for the coming months.

Jackie James
ISMRD Board President
Mom to Anna, ML III

ISMRD's Mucolipidosis Research Initiative

The Wagner Foundation

Update on ISMRD's Mucolipidosis Research Initiative

Following the fabulous efforts of ISMRD families, friends and supporters, and with the great support of our partner organizations to raise $150,000 for research into Mucolipidosis, we made a call for Letters of Interest from researchers to use these funds.

As this edition goes to press, we can report that six high quality proposals were received from researchers in USA and Europe. Sadly we cannot fund all of them. Over $400,000 would be needed to do that.

A committee formed by the Chair of our Scientific Advisory Board, Steve Walkley, has evaluated the bids and short-listed three of them. We will ask those three researchers to submit a full proposal soon, and recommendations as to the successful bidder(s) will be considered over November.

The successful bidders will receive funds in mid-December, so the funds you put so much great effort into raising during the first half of this year, will be allocated and put to work before the year is out.

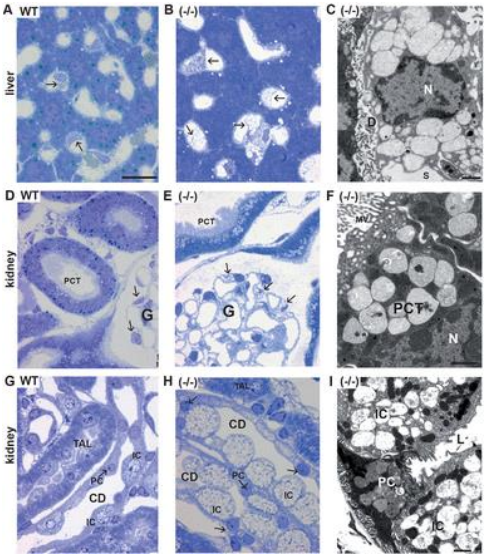
This is a great effort from all of you. Thank you so much.

Advance in Fucosidosis Research

Fucosidosis is one of the rarest of ISMRD's nine glycoprotein storage disorders. As a result, it tends to attract less research than many of the other disorders. This makes some new research on Fucosidosis all the more exciting.

Research by Heike Wolf, Markus Damme, Stijn Stroobants, Rudi D'Hooge, Hans Christian Beck, Irm Hermans-Borgmeyer, Renate Lüllmann-Rauch, Thomas Dierks and Torben Lübke has resulted in the generation of a Fucosidosis mouse model, which resembles the human disease.

The mouse model is an easy to manage system in order to understand the mechanisms of disease progression, to identify putative biomarkers for reliable diagnosis and to address therapeutic strategies such as Enzyme Replacement Therapy (ERT). Because of their small size, mice would only require very small doses of the very expensive recombinant fucosidase enzyme.



Histopathology of liver and kidney in 5-month-old *Fuca1*-deficient mice

Having this mouse model will help to unravel underlying pathological processes, and could be used to establish diagnostic and therapeutic strategies for Fucosidosis.

Lysosomal storage pathology was observed in many visceral organs, such as in the liver, kidney, spleen and bladder as well is in the central nervous system. A progressive loss of Purkinje cells combined with astrogliosis led to psychomotor and memory deficits.

The researchers are now going to begin ERT trials with the mice.

For the full paper, go to A Mouse Model for Fucosidosis

Shamim, Edward, Shirley and Sam

Mucolipidosis wristbands raise awareness and funds

My son Sam's school special needs coordinator, Angela Kearney, noticed the reminders coming up on my Facebook page for Rare Diseases Day on May 15th. She asked me if it would be ok if the school purchased wristbands with the message 'Mucolipidosis Awareness' on them and sold them in school to raise awareness and funds. The school ordered 500 and charged UK£1.00 for each wristband. We were quite touched by the suggestion and the wristbands sold very quickly.

Our local church, Sacred Heart-Rochdale, where Sam is an altar server, also asked if they could help raise awareness by selling the wristbands. Approximately 100 were sold in the space of 24 hours, and donations reached £200.

Approximately £420 was raised altogether, and split between the UK MPS Society and the ISMRD who both support our family.

Over the years, people have seen Sam limping or in his wheelchair. They sometimes ask "What is wrong with him?". To be honest, we can't always be bothered to tell them the full story as you lose them when you say 'Muco'. When they see the wristband, they can look it up. I'm not saying they'll understand it, but at least they will have seen the name.

The aim is to raise awareness, and hopefully gain some support and maybe research in to a cure for Mucolipidosis. Dare to dream!

As an added bit of fun, we decided to send the wristbands to interesting places around the world, and places such as the theatre, seaside, pub. Anywhere where you'd be proud to take a photo of it and share it on social media. Therefore, spreading the awareness even further.

We used the strapline "Mucolipidosis - A Cure starts with Awareness"'

So far the wristbands have been tagged in Barbados, Dubai, Malaysia, Honduras, Hollywood, Rome, Spain, Venice, New Zealand, Wales, London plus many more cities. They've been abseiling and skydiving, and also seem to like visiting bars, music and sports events too.

Shirley Jamil

ISMRD PENGUIN BRACELET

Who can resist this beautiful bracelet? With a delightful glass penguin pendant, faux pearl beads and letter beads spelling out "ISMRD", this bracelet stretches to fit most wrists. Buy one for yourself, ask your friends to sell them. Only twenty dollars each, they are adorable and very good awareness-raisers. Go to our store at www.ismrd.org/fundraisers/ismrd-shop

Only $20 each

Amazon Smile

If you shop at Amazon Smile, a portion of the purchase price will be donated to the ISMRD, at no cost to you. **Y**ou'll find the same low prices, vast selection and convenient shopping experience as Amazon.com.

Go to http://smile.amazon.com for this easy way to help raise funds for the ISMRD.

Exciting opportunity for ISMRD members

The nine ISMRD glycoprotein storage disorders have been added to the ConnectMPS registry, a centralised comprehensive global database that connects families, researchers, industry and all stakeholders in the MPS and ISMRD communities . This means that ISMRD family members can enter information about their affected person or people into the world's first pan- mucopolysaccharidosis online patient registry. Entered through a secure website, the de-identified data are then shared with research investigators and companies working to develop new treatments.

The registry program includes 22 national and international advocacy partners through a partnership with the National MPS Society.

Known as ConnectMPS, it is designed to better understand the medical history and impact of the included diseases by collecting information directly from the patient community.

By coming together, we as affected families not only demonstrate our unity towards a cure but offer researchers and industry the most efficient and uniform source for patient data to advance all treatments.

This program is the first-of-its-kind online patient centred registry. Researchers and drug developers alike can better understand the progression of MPS and ISMRD diseases, available through a central, openly - accessible portal. The registry provides a secure portal for patients to contribute, you and share data, while safeguarding their privacy.

Participants can also receive information about the upcoming clinical trials most relevant to them.

For more information, go to ConnectMPS

To join, go to Registry

If you have any questions, please contact: connect@patientcrossroads.com

What's happening around the world?

State of rare disease management in Southeast Asia

An article published in the *Orphanet Journal of Rare Diseases* consists of information from six Southeast Asian countries on management of rare diseases based on the World Health Organization's framework for action in strengthening health systems. The authors conclude that even through there is some progress, *"rare disease management remains challenging across Southeast Asia".* They also compare the facilities in these countries to France, Canada, Australia, Taiwan, and South Korea, which they consider to follow best practices in rare disease management. According to the authors, *"Southeast Asian countries have made significant progress in the management of rare disease, but there remain key areas for substantial development opportunities."*

State of Rare Disease Management in Southeast

Newborn screening for Krabbe's disease in the US

Live newborn screening for Krabbe's disease (KD) was initiated in New York in 2006, and started in Missouri in 2012. As of August 7, 2015, nearly 2.5 million infants had been screened, and five infants had been determined to have KD. As of August, 2015, the combined incidence of infantile KD in New York and Missouri is ~1 per 500,000; however, patients who develop later-onset forms of KD may still emerge. Although current treatment options for infants with early-infantile Krabbe's disease are not curative, over time treatment options should improve.

Parenting a child with metabolic diseases: Impact on health related quality of life of parents

The health related quality of life (HRQoL) of the parents of children with inborn errors of metabolism was assessed among the outpatient population in Sultan Qaboos University Hospital, Oman. Published in *Diabetes & Metabolic Syndrome: Clinical Research & Reviews*, this study showed that parents with higher educational qualifications, higher monthly income and younger age had a higher HRQoL.

Additionally, a strong support system and help towards preparing special diets for the child also played a role in predicting the parents HRQoL.

EURORDIS Photo Contest 2016

Open to anyone with an interest in rare diseases. You have until 5 December to send your most beautiful, original, or artistic photo(s) to try to win an iPad and other prizes.

There are 3 categories in the EURORDIS Photo Contest: **Public Prize**, **Expert's Choice**, and the **Instagram Prize.** Find out more about our judges.

Enter by filling in the form at photo contest form or on Instagram with #RareButReal2016.

Emory Genetics Laboratory created the "My *Beautiful* CHILD" social media campaign to openly communicate and to create greater awareness about chromosomal abnormalities and other genetic disorders seen in children.

"My *Beautiful* CHILD" is a collection of personal photos and narratives, celebrating real children and families leading lives a little different from others. The program seeks to connect those impacted by genetic disorders seen in children, share information and resources, and promote a sense of positive community. This is made possible by children, their families, caregivers, and clinicians; advocates; and others in the medical and scientific community coming together to re-establish a fundamental message: every child is a beautiful child.

You are invited to join this online community by submitting your pictures and story at My Beautiful CHILD. Each month, new families will be spotlighted and resources provided to further network and educate the community. There is strength in numbers and while genetic disorders may be individually rare, those impacted should never feel alone.

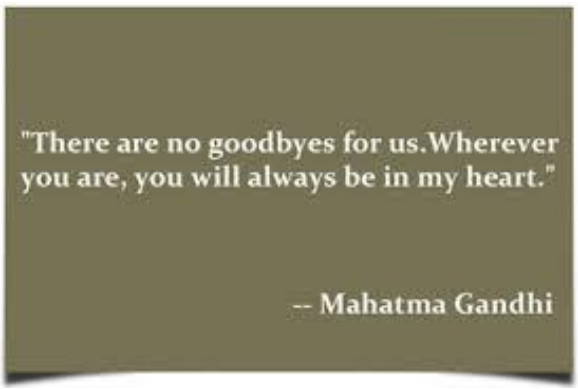
Learn more and participate at My Beautiful CHILD.

We mourn the passing of Tatym Stapf, who died on 5 August 2016, aged 16 months.

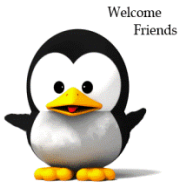
Tatym's Fucosidosis was being treated with a Bone Marrow Transplant. She died as a result of complications of the Bone Marrow Transplant.

She lived in South Dakota USA.

Our deepest condolences to her family, including her sister Aiva who is two years and 10 months old, and who also has Fucosidosis.



ISMRD WARMLY WELCOMES YOU TO OUR FAMILY



- Franziska Huber is from Austria and has Mucolipidosis ll. We also welcome her family member Evelyn Huber

- Ashwarya and Udit Utkosh, also a Mucolipidosis ll family, live in Chicago, US

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

- Sergio Cardenas, who has ML ll/lll, went into Texas Children's Hospital with fainting, shortness of breath, chest pain and tiredness relating to his heart. He has also had hip injections under general anaesthetic
- Fardowsa, who is 19 years old and has ML lll, had surgery in September for a spinal tethered cord release
- Ivan Garcia, who has Galactosialidosis, had surgery to help with his asthma
- Luci Kesner is 4 years old and has ML ll. She is in hospital with pneumonia
- Malynda Norstadt, who has recently had surgery

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

Mary Kimmet and
Martin Woolley

for their very generous donations

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