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

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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

As the new President for ISMRD it gives me great pleasure to announce ISMRD's future direction for supporting research for our diseases.

In July this year we concluded a very successful conference in St Louis. This was the fifth workshop/conference organised by ISMRD since our formation in 1999 and this highlights the impact we have had in generating interest in research into our nine diseases. Research is a top priority issue for ISMRD, alongside our other key priority of providing information and support to our families.

Over the last 10 years, ISMRD has raised in excess of $500,000 to support research. This includes funds raised for our conferences, the Natural History Study and other specific projects, and includes funds that came from the hard work of our families, plus sources such as the National Institutes of Health (NIH), various Mucopolysaccharidosis (MPS) Societies, Industry and other Lysosomal support groups to pool our funds.

What we have achieved:

- One family/medical workshop and four International family/scientific conferences.
- The Natural History study for our diseases at the Greenwood Genetic Center, with support from the NIH, Lysosomal Diseases New Zealand, and the US MPS Society.
- A Bone Consensus meeting held in New Zealand, with support from the UK MPS Society and Lysosomal Diseases New Zealand.
- A $40,000 grant, in partnership with the US MPS Society, to Heather Flanagan-Steet at the Complex Carbohydrate Research Center at the University of Georgia, for research on the role of cathepsin proteases in ML-II cardiac pathology.
- A Patient Advocacy Leadership award from Genzyme to support the development of Mucolipidosis Clinical guidelines.

We are currently actively exploring other research initiatives such as read-through therapies for nonsense mutations and further evaluation of bone disease management options for our diseases.

So how do we move forward all this and other projects relating to research and what is our priority for the next year?

At our last board meeting the Board of Directors noted that Mucolipidosis is very complex and is probably the hardest of all the Lysosomal diseases to solve. It also languishes among the least researched of all Lysosomal diseases.

We have some funds raised specifically for Mucolipidosis research and we have decided to join with other support groups and foundations, to produce a larger pool of grant funding thus allowing for more significant research to take place.

The Board of ISMRD has set aside $30,000 to kick-start the Mucolipidosis research fund. This is where you, our families, can help us be successful in raising money for Mucolipidosis research.

The contributions received from family fundraisers and donations have been a vital part of these research efforts, and we are now calling on your help so we can do more.

ISMRD has $30,000 in the pot. What can you add to this? $100,000 is our goal.

In this newsletter is an article publicising this project. We are asking for your help to reach our target of $100,000 by June 2016. What can you do to help?

This special fundraising target will run alongside the need for funds for our other general needs, and of course we will continue to accept funds that may be focussed on the needs of the rest of our nine diseases. All donations are most welcome.

We have recently created a Fundraising Ideas page and very shortly we will have a merchandise page up and running, selling various items such as rubber bracelets, Penguin bracelets, ISMRD tee-shirts and our very own blend of tea.

Rare Disease day is just around the corner – What a fantastic opportunity for you all to get out and raise awareness and raise funds for our diseases.

You have probably noticed the fantastic new Facebook pages we have recently set up for each individual disorder that ISMRD covers. Jenny Noble has done a wonderful job of putting these pages together for our families and friends to be able to interact with one another and draw in new families searching for support and information.

ISMRD is happy to announce that Juanita Van Dam from Australia will be stepping up to the plate as a new Board member. Welcome Juanita! We very much look forward to have you working with us in 2016!

At the same time we are sad to see Susan Kester step down from the Board – but pleased to say that she has agreed to retain her position heading up the "Sunshine Committee" that takes care of keeping in touch with those that are dealing with surgeries, family issues and the loss of loved ones. All that know Susan would agree that she is indeed a ray of sunshine and has been an invaluable asset to the ISMRD Board. Thank you for all that you do Susan.

During this busy season, we remember those that have gained their angel wings, along with those that have had or are about to have surgery. Our thoughts and prayers are with each of you.

Wishing you and your family all the very best for the festive season,

Jackie James
President, ISMRD

Support ISMRD's exciting Mucolipidosis Research Initiative

ISMRD has set aside $30,000 for Mucolipidosis research. Our goal is to amass a total of $100,000 for Mucolipidosis research by June 2016.

What can you contribute to this?

Mucolipidosis Alpha/Beta and Mucolipidosis Gamma are ultra-rare and very complex conditions. Among the many Lysosomal diseases, they stand out as more complex to understand and more challenging to develop therapies for.

Their problems are not "standard" enzyme deficiencies with resulting storage bodies. They have more obscure problems related to trafficking of enzymes to the Lysosome. They are less understood. They are less researched. We intend to change that.

We are seeking funds from our families and supporters to add to our campaign pot. We will also partner with other support groups around the world to make the $100,000 target achievable.

Our goal is $100,000 by June 2016, and a call for research proposals in the second half of 2016.

ISMRD has a proud history of initiating research on our diseases. This campaign will step things up to another level by enabling more significant research projects to occur.

We believe this is achievable, but we need your help.

Let's work together and get research happening that will give greater knowledge, understanding and one day a viable therapy for the Mucolipidoses.

Watch this space for donation details in the new year.

Andre, Sergio, Zachie and Aiobhe,
all affected by Mucolipidosis

URGENTLY NEEDED!

Person with Social Media Knowledge

ISMRD needs to get its message out to the world. Communication technology keeps evolving and we need a savvy person to help us with Twitter, Instagram and Facebook.

We would appreciate it if you would monitor these for us, and provide ongoing advice and assistance to help us lift the profile of ISMRD.

Board members can write the messages if you can "post" them for us.

Please contact info@ismrd.org

NEW: a Facebook page for each of our disorders

ISMRD is developing new information pages on Facebook for each of the nine diseases that it supports. The ISMRD group Facebook page is closed and private, and is for chatting, venting, possibly sharing personal details about our family members. We will keep this page going. The new individual information pages will carry announcements, latest research, events such as Rare Disease day and other such announcements. They are open to the public so you may not want to post personal information on there. Feel free to post events, fundraising etc.

We intend to lift our profile with these individual Facebook pages. We will also be putting our diseases into RareConnect, which is similar to Facebook, but which will allow us to reach out to Europe where we know there are many more families we have not yet been able to connect with.

For more information on RareConnect, go to: https://www.rareconnect.org/en

Here are the links for our public Facebook pages:

Mucolipidosis Alpha/Beta and Gamma	Mucolipidosis Alpha/Beta and Gamma Facebook page
Fucosidosis	Fucosidosis Facebook page
Mannosidosis Alpha and Beta	Mannosidosis Facebook page
Galactosialidosis	Galactosialidosis Facebook page
Aspartylglucosaminuria (does anyone have any photos?)	Aspartylglucosaminuria Facebook page
Schindler Disease (does anyone have any photos?)	Schindler Disease Facebook page
Sialidosis	Sialidosis Facebook page

Please visit these pages, "Like" them, Share them with your Friends

New ISMRD Fundraising webpage

ISMRD is able to hold its international conferences, provide family support and information and promote research into our glycoprotein storage disorders thanks to funds raised by individual ISMRD members. Without these funds raised by you our members, our activities would grind to a halt.

Are you doing your bit?

Go to http://www.ismrd.org/fundraisers

to see our exciting suggestions for how you can help the ISMRD continue its valuable activities.

More about this in future issues.

Don't forget the ISMRD gofundme page, which has so far raised US$4,501. Every little bit helps. If you would like to donate, go to http://www.gofundme.com/5rpjhw

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. You'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.

What's happening around the world?

Finally: some evidence that enzyme replacement therapy saves money!

Cost effectiveness of enzyme replacement therapy for Mucopolysaccharidosis: a Brazilian Study

An article published in *Value in Health – Regional Issues* evaluated the frequency of medical interventions in a cohort of patients with mucopolysaccharidosis (MPS) I, II, and VI, which are lysosomal storage disorders on enzyme replacement therapy (ERT) to estimate the impact of direct medical costs associated with the treatment of MPS and compare its frequency with that observed among patients not on ERT in Brazil.

The authors collected data on the number and type of medical appointments, hospital admissions, medications used, and surgical procedures performed per patient through a review of medical records, as were data on ERT. The authors found that overall hospital admissions and surgical procedures per year were higher in the non-ERT group. Thus they concluded that excluding the cost of recombinant enzymes, Brazilian patients with MPS I, II, and VI who receive ERTs undergo fewer medical interventions than do patients in supportive care.

Read more: Cost Effectiveness of ERT

EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS

Edinburgh, UK, 26 – 28 May 2016

Read more: Conference Program

European Conference on Rare Diseases & Orphan Products

Pernille Roll (ML lll) celebrates Halloween in Norway

Carer Resources

Genzyme Blog Series on
Building Resilience for Rare Disease Caregivers

Vanessa King is an expert on positive psychology and resilience. As part of a blog series developed by Genzyme to help rare disease carers build resilience, she talks here about resilient thinking, and resting well.

Resilient Thinking

As patient group leaders, we can often feel under pressure - especially so when things don't always go to plan! When this happens, what are your typical thoughts about why that problem occurred? Do you generally jump to what you did that might have caused it or feel that it's typical – these things always happen to you? If so, you could be undermining your own resilience.

Science is showing that the way we interpret the events that happen day-to-day in our lives has a significant relationship to our ability to cope and deal effectively with problems, and on our physical health and persistence in the face of adversity.

So which ways of interpreting these events are the most resilient?

Let's look at three factors:

1. Is it me? - When bad things happen, resilient thinkers tend to focus on causes outside of themselves. For example if they are late they will look at the delay on the trains or heavy traffic as being the main cause of the issue rather than getting stuck on beating themselves up for not leaving the house earlier.

2. How long will this problem last? - When things go wrong, resilient thinkers see it as transitory, perhaps thinking "It didn't work this time, but next time it will be better." In contrast, someone with a thinking style

that isn't resilient might think it will always be that way, e.g. "It didn't work this time, and it's never going to work."

3. How many different aspects of my life will this affect? – When something goes wrong in one area of a resilient thinker's life, they put boundaries around the issue limiting it to that specific area, for example: "I took the wrong turn, I find map reading hard." Whereas someone with a non-resilient way of thinking will see the problem as spreading out to everything, e.g. "I took the wrong turn, I'm no good at anything."

It isn't about being unrealistic or just kidding ourselves when day-to-day problems occur. It's about being realistic and flexible in our thoughts about why these issues happened. If we are tired and stressed, we can all too easily fall into the trap of thinking we are the cause of all things that go wrong, that problems will always be that way and that they spread out to everything.

So next time something goes wrong for you. Pause for a moment and think realistically: How did I, others or the situation contribute to this? What can I do (or ask of others) now or that will help stop the problem occurring again? What strengths do I have that I can draw on to help? (We'll look at strengths in more detail in an upcoming blog). More resilient patterns of thinking really can be learned.

You may also find it helpful to have a look at the attached guide called "Check Your Thinking" which can help us manage our emotional responses.

Resting well

Sleep matters. Scientists suggest that we should sleep for at least one hour for every two that we are awake. That adds up to 8 hours a day.

Yet we live in a society where most of us are chronically sleep deprived – we lead busy lives at home and at work, are constantly connected to our phone, laptops or TVs. If we are carers, we may have even greater challenges.

Take sleeping seriously

People who sleep sufficiently are generally happier, healthier and have better cognitive functioning than those who get too little. So what can we do to sleep longer and better? The first thing is to take sleep seriously – it really does make a difference to how we feel and function and our longer-term psychological and physical health. We'll look at some tips to help you get to sleep and sleep well for longer.

Sleep experts recommend trying our best to have a regular time for going to bed and waking up. As carers, this isn't always easy to do as we often have to get up to attend to our loved ones during the night. However, there are no rules saying that we need to sleep for our 7 or 8 hours straight through. In fact our ancestors often slept for 4 or so hours (from say 8:00 pm to midnight) then got up for a few hours spending that time in thoughtful reflection or connecting with others, then going back to bed for a "second sleep" in the early hours. So if you do have to get up in the night, can you arrange to have a nap during the day?

Tips on sleeping well

• **Lower the lights** – we are biologically programmed to respond to the natural cycle of daylight and darkness. The pineal gland behind our eyes responds to the level of light by switching on or off our production of melatonin, which governs our sleep. We now spend a lot of our time in artificial light, which confuses our natural system so a simple act of dimming our overhead lights in our sitting room and bedroom a couple of hours before going to bed will help.

• **Tame technology** – as tempting as it is to be glued to our screens late into the night, it is a huge disrupter of sleep. Yes it may help us relax and switch off, but as far as sleep goes it switches us on. Our TVs, phones and computers all emit a blue light of specific wavelengths that really mess with our production of melatonin and hence our sleep. So turn off your screens 1.5 to 2 hours before you go to bed. Or if you really must use the time before bed to catch up on emails etc., investigate apps or software that change the quality of light from your device or invest in a pair of amber lensed glasses – they may not look great, but you'll look and feel better the next morning!

• **Limit late-night liquids** – a full bladder is a common cause of restlessness or waking up in the night. So make sure you drink plenty of water and other fluids during the day and stop at least 1.5 hours before you go to bed. (Alcohol included – we may feel it helps us get to sleep, but it disrupts our naturally patterns causing us to wake in the early hours).

• **Neutralise noise** – often we feel that we should eliminate all noise from our bedrooms. However, centuries ago, noise was a key factor that helped people sleep! But it's the type of noise that matters. If you can hear noise from outside that bothers you then spending a few pounds, euros or dollars on some foam earplugs could really help (and are very good if you are travelling).

In contrast, gentle sounds that signal comfort and safety can actually help us sleep better than no noise at all. For example, the sound of a pet sleeping, the radio tuned to a talk show at low volume, a recording of gentle waves or the gentle whirr of a fan all can work. So this might be worth a try.

• **Switch off your thoughts** – sometimes what stops us going to sleep is our minds. They just won't stop whirring, perhaps stuck on a problem you have to deal with, overly full to do lists or worrying. If this is the case for you there are some simple tricks you can try – have a look at the attached guide for some suggestions and find the one that works best for you.

If you give these tips a try and you still have problems, do make time to speak to your doctor. Your sleep is too important not to!

Genes in Life

Family Health History Toolkit

The US Genes in Life organisation has developed a Family Health History toolkit which you can download at the following link. People in the US can also ask for a hard copy to be mailed to them:
Family Health History Toolkit

ISMRD warmly welcomes

to our family

- William Cochran and family. William has Alpha-Mannosidosis, and lives in Auckland, New Zealand. He is undergoing a transplant.
- Ryan Dernie and family. Ryan has Alpha-Mannosidosis, and is 14 years old. He had a transplant 10 years ago and is doing well. He and his family live in Auckland, New Zealand.
- Aiva and Tatym Stapf and family. Aiva (2) and Tatym (7 months)have Fucosidosis. They live in South Dakota, USA.
- Mojca Jamnik, who has a family member with Mucolipidosis
- Melissa McCarty Statham, Otolaryngologist at Atlanta Children's ENT, Georgia USA

May the stars carry your sadness away,
May the flowers fill your heart with beauty,
May hope forever wipe away your tears,

Sadly we mourn the loss of Lukas Maciukeviciute. Lukas was 19 years old and had Mucolipidosis lll. He and his family recently joined the ISMRD, and live in Lithuania. Our hearts go out to Lukas's family.

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

- Heather Scott, who has Mucolipidosis lll, has been unwell and in hospital
- Robert Stark, who is 19 and has Alpha Mannosidosis, had hip surgery (Periacetabular Osteotomy or PAO) in October
- Skylar Thomas, who is 22 years old, has Mucolipidosis III and lives in Austin, Texas, had a knee replacement in October
- Anne-Pia Salo from Finland is 49 years old and has Alpha Mannosidosis. She is very ill.

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

- David Dollins
- Shirley Jamil
- Mary Kimmet
- Kelly & Jamie Moran
- Diane Sanders
- Mark Stark
- Martin Woolley