

# Pathways

## The Newsletter for Glycoprotein Storage Diseases

ISMRD, a 501 © not-for-profit organization, FEIN 53-2164838 | website [www.ismrd.org](http://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, Mucopolipidosis II alpha/beta (I-Cell Disease), Mucopolipidosis III alpha/beta (Pseudo-Hurler Polydystrophy), Mucopolipidosis III Gamma, Schindler Disease and Sialidosis

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**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



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## From the President's Desk

*By Jackie James  
President, ISMRD*

It is with great pleasure that I write my very first greeting to each of you as ISMRD's new board president! I am mom to Anna, who has ML III and is now 20 years old. I am originally from the UK but moved to St Louis, USA many years ago where I live with my husband and two children. I have been an ISMRD board member for the past four years and I am so excited and honoured to be named ISMRD's new board president.

It's now been over a month since we all gathered together for the ISMRD 2015 family and scientific conference here in St. Louis. What a wonderful time we had as we caught up with old friends and met new ones for the first time. We have some wonderful reports from Carolyn Paisley-Dew, John Forman and Sylvia Webb included in this publication for you to enjoy. Thank you to each of you that came and made this conference so special.

Please join me in extending a very big welcome to our new families! We are so happy to have you join us. At the same time, there are several of our kids that have either just had surgery or are about to, and I'm sure you will join me in wishing them a successful outcome. Details of new families and surgeries are listed in this newsletter.



This newsletter also includes some wonderful information on the new Croatian National program for rare diseases. It is so great to see more and more countries develop these programs!

ISMRD is very excited that we have been accepted as a member of NORD (US National Organization for Rare Disorders). As a member, this allows us to become part of a large collective voice on behalf of patients with rare diseases.

I've decided that I need to make a trip to San Antonio, Texas to visit Morgan's Wonderland accessible amusement park! Please visit our special "Carer" page to get more details. It sounds fantastic!

Please make a point of reading the article by Vanessa King (Genzyme) that gives invaluable insight for each of us that are caregivers for our children. I'm sure you will agree that it is very easy to forget to take care of ourselves when we are primary caregivers for our kids.

Lastly I would like to thank each of you that have contributed financially to ISMRD this year. Your support is invaluable.

I wish each of you much happiness and good health

**Jackie James  
President  
ISMRD**

ISMRD's **MEET US  
IN ST. LOUIS**

**FOR THE 4TH  
INTERNATIONAL CONFERENCE  
ON GLYCOPROTEINOSES**

Washington University in St. Louis  
SCHOOL OF MEDICINE

*Crossing oceans for a cure*

JULY 23 - 26TH 2015 VENUE: HILTON AT THE BALLPARK, ST. LOUIS, MISSOURI, USA



## Glycoproteinoses: Fourth International Conference on Advances in Pathogenesis and Therapy



The fourth International Conference hosted by ISMRD was the biggest conference ever. More than 200 people milled around the halls of the St Louis Hilton at the ballpark. Almost all of our nine disorders were represented by an affected family. Speakers gave presentations on all of our disorders. People came from 15 countries and four continents.

We all learnt some more about our specific disorders and the latest advances in research and therapy for them.







We met as one big international family and rejoiced in each other's company.



The children had a wonderful time. They went to the Zoo ,



had Pampering Sessions



hung out



Darth Vader, Anna from Frozen and many other celebrities visited.





A moving Memorial Ceremony was held in the Ballpark next door to commemorate those that have died of a Glycoproteinosis.



The Gala Dinner was lavish, and all our hard-working presenters received a thank you gift.



Mark Haskins, from the University of Pennsylvania, received an award from the ISMRD for his long-term commitment to Lysosomal Storage Disorders.





Paul Murphy also received a Life Time Award for his pioneering work in matching the needs of rare diseases with the internet age and enabling the development of ISMRD

**Carolyn Paisley-Dew**  
**Board Member, Editor**

The Board members met face-to-face for the first time since the ISMRD Conference in South Carolina in 2012.



Everyone visited the St Louis Gateway Arch, which opened in 1965. It is 630 feet (192 metres) tall and has trams to take you to the lookout at the top.

**Thank you, St Louis!**

## FAMILY WORKSHOPS



John Forman, ISMRD's Vice President, Research, attended the conference in St Louis with his son Tim. Friday afternoon and Saturday morning saw separate workshops for family members. There was one for Mucopolidosis, which has the greatest number of affected people of all the Glycoproteinoses, and a separate workshop for the other disorders (Alpha Mannosidosis, Beta Mannosidosis, Fucosidosis, Sialidosis, Galactosialidosis, Aspartylglucosaminuria and Schindler Disease).

Here John reports on this second workshop, which he attended as Tim has Alpha Mannosidosis.

This workshop included very informative and interesting talks on neurological symptoms of our diseases, a summary of knowledge of Fucosidosis, experiences with cell transplants, new developments in therapies, and discussion of education issues. A notable theme in the discussions related to the advances in understanding and managing many symptoms of the diseases, and also the great momentum that is occurring in development of therapies. There are many different therapeutic options being explored, with many of them now in or very close to clinical trial phase.

The workshop was run with a nice intimate feel to it, enabling families to ask questions and talk to the presenters in a nice and relaxed atmosphere, away from the more formal setting of the main scientific presentations. This added to the good feelings derived from the positive discussions about emerging therapies, which was a major theme of the whole conference.



The St Louis Cardinals play the Atlanta Braves





## Report from Mucopolidosis II/III Parent Sylvia Webb

One of the motivations behind us attending this Conference was to reunite Grace with her 'brother from another mother', Zach, who we met 3 years earlier in Charleston, South Carolina. My husband Charles and I attended this Conference with our two daughters affected by Mucopolidosis II/III, Tegan and Grace, our son Christian and our daughter Caryl along with her two sons, Liam and Chase.

During the Conference, I attended most of the presentations while Charles, Christian and Caryl acted as carers to the young people attending the Children's Program of which Tegan and Grace attended the 'girls' and combined activities while Liam and Chase attended the 'boys' and combined activities.

So many people came to the conference. All came for their own reasons. Some came to give information. Some came to get information. Some came to catch up with friends they had met at other Conferences/meetings and/or make new friends. Some came to care for the young ones while on outings or participating in recreation activities while their parents attended Conference presentations.

Being summer in the USA, the weather was hot. The hotel however provided excellent air conditioning to keep delegates comfortable while attending presentations. The hotel and ISMRD also provided delicious breakfasts each morning of the Conference and a lovely meal during the Gala Dinner on Saturday night.

### Combined Scientific and Family Conference Presentations

The Conference started with a Welcome Reception on the Thursday night before it began in earnest the following morning. The conference was divided between Professional and Family presentations with some overlap particularly where subjects presented by professionals targeted specific diseases. As our children have a Mucopolidosis disease, I attended the relevant scientific presentations highlighting the information pertaining to Mucopolidosis.

Some of the professional presentations detailed work being carried out on animals born with the same in-born errors of metabolism as our children. Many were very technical with presenters using scientific jargon which often left me and others feeling more confused at the end of the presentation than when the speaker started. Luckily, most of the presentations included slides or pictures which sometimes helped.

The bottom line from all of these presentations is that there is no treatment or cure for our children anywhere in the near future. The view of the scientific field, at the moment, is that children with Mucopolidosis II, II/III or III will, in the future, probably need a combination of several treatments or therapies to modify or correct their metabolism.

The Family Presentations were more about the various side effects or complications of the diseases and the treatments of those. Some of these topics included:

- Surgical treatment of spinal cord compression
- Heart issues, in particular, infiltration of heart valves causing regurgitation of blood between the chambers
- Schooling issues
- Obsessive Compulsive Disorder and Grief.

The common thread throughout these presentations was the limited amount of data available to describe many of these complications and the treatments attempted to improve the comfort of the children and young adults suffering these complications.

While the information presented regarding the spinal cord compression and heart issues is not new, hearing it and putting it into practice is often the challenge for families. The speakers who have experience are often from centres far away from the families needing the treatment for their children. In my own experience, many doctors can be quite arrogant and not want to accept the word of a parent, especially where the parent may be emotional when describing their child's symptoms or frustrated at the lack of empathy on the part of the treating doctor or specialist.

The schooling issues presentation centred around what parents need to know to be the best educational advocate for their child/ren. Topics included:

- How to decide which skills your child needs to learn
- What kind of help and environmental supports your child may need in order for effective learning to happen
- Ways to support your child to help maximise functional learning.

While this information was very informative, in my experience, it can be difficult to put these things into practice, depending on the resources of the school, how receptive the staff are to new information and how proactive they are in putting ideas raised into practice. In Australia where we live, where special schools are not always available and integration within mainstream schools may cause hardship, not only for the disabled child but the other students and the teacher, parents may face battle after battle before they see their child being supported in a way that suits them and their needs.

The discussion about Obsessive Compulsive Disorder (OCD) generated many nods and smiles. It seemed that many of the parents attending had children who displayed traits of OCD. I raised the notion that considering that all of these children had little to no control over most aspects of their lives, especially in relation to i) having the disease and ii) coping with the pain and complications of the disease, that perhaps OCD was a way for them to 'be in control' of some aspect of their lives.

One of the things that came from the discussion was the positive aspect of partnering with other parents to discuss and seek advice when new or interesting signs or symptoms became evident in their child's life.

### **The Children's Program**



On the Friday, all the children and carers went to the St Louis Zoo. This zoo, as well as many other tourist attractions in St Louis is free for the public to attend. Charles told me it was the best zoo he had ever been to and we've been to six different zoos in Australia.

On the Saturday, during the day, the younger children had art therapy and storytelling. The older girls enjoyed beauty therapy/pampering with hair styling, make-up, nails as well as a posh morning tea. And the older boys went over the road from the hotel to tour the St Louis Cardinals Baseball Stadium. St Louis love their Baseball team, The Cardinals, and support them ferociously. While we were in St Louis, there was a baseball match almost every day/night.



In the evening the Conference Foyer was decorated to resemble a Circus tent and some interesting performers came and entertained the young people and anyone else interested in joining in the festivities.

On the Sunday, Darth Vader and his friends visited and many of the children and carers had photos taken with their favourite character.

### **Meeting Families**

For many of the families, including us, I believe that the children were the biggest draw. Three years ago when we attended the Conference in Charleston, South Carolina, we met two young boys with the same strain of Mucopolidosis that our children suffer from: Zach from Syracuse, New York and Sergio from Houston, Texas. Zach in particular became Grace's 'brother from another mother'. This year we met more but the eight year old girl from Ireland, Aoibhe (pronounced Ava) stole everyone's hearts. Now the girls have a new 'sister from another mister'. Grace would get quite jealous when Aoibhe sat next to Zach.



Mucopolidosis is such a rare disease. To our knowledge Tegan (32 this year) is the oldest young person in the world with her strain of the disease. Other parents with disabled children can relate to each other and the challenges they each face but another mother of a Mucopolidosis child not only relates but becomes related in the family sense.

### **Memorial Service**



Prior to the Conference, family members of ISMRD were asked if they would like any of their children who had died to be remembered during the Memorial Service. On the Friday, a Memorial Service was held in memory of these children and young adults who have died from complications of the various diseases. Doves were released as part of this Service as the family members in attendance watched.

Sadly, many family members including children were on an outing to the zoo and were not able to attend.

### **Natural History Study**

In 2009, ISMRD funded a project to document the natural progress of the various Mucopolidosis diseases. At the 2012 Conference in Charleston, South Carolina, a medical clinic was held at the Greenwood Genetic Centre so that all young people who were part of the Natural History Study could meet with medical specialists and have various specimens taken so as to gather more information. A formal clinic could not be held at the St Louis Conference due to lack of funds and resources but Dr Sara Cathey from the Greenwood Genetic Centre in Charleston, South Carolina was still able to collect blood and urine sample and have them shipped back to the Greenwood Genetic Centre for analysis as part of the continuing Natural History Study.



**Conclusion**

I've always enjoyed Family Conferences and learned new things each time we've attended. I have always especially valued the connections made with other families. The 2015 Conference in St Louis was no exception. Now we know more families with ML II, II/III and III from around the world and it's wonderful to know that my 'family' is growing.



**ISMRD is very grateful for the very generous support of the following sponsors for the Fourth International Conference on Glycoprotein Storage Disorders:**

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Don't forget the ISMRD gofundme page, which has so far raised US\$4,451. Every little bit helps. If you would like to donate, go to <http://www.gofundme.com/5rpihw>

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## What's happening around the world?



The Croatian National Programme for Rare Diseases was approved by the Croatian government in February this year. The Programme represents nine strategic areas of activity to be fulfilled during the period 2014-2019.

- Improving knowledge and access to information on rare diseases (RD) in Croatia
- Supporting the development of registers of RD and their permanent financing
- Supporting the development of the network of reference centres and centres of excellence for RD
- Improving the availability and quality of health care (diagnosis, treatment and prevention)
- Ensuring the availability of drugs for RD
- Expansion of social rights of people affected by RD
- Empowering patient organisation
- Promoting scientific research in the field of RD
- International networking and cooperation in the field of RD

[Read the Croatian National Programme for Rare Diseases](#)







# Carer Resources



## Morgan's Wonderland, San Antonio Texas, USA

Morgan's Wonderland is a 25 acres accessible amusement park in San Antonio, Texas, USA for children and adults with special cognitive and physical needs.

Entry for people of any age with special needs is FREE. \$17 for carers and adults. Discount rate for special needs groups (10 or more paying guests) is \$9 per person.

"Our vision at Morgan's Wonderland is to establish a special place where smiles and laughter make wonderful memories with family members, caregivers and friends. To build a place where the common element of play creates an atmosphere of inclusion for those with and without disabilities, encouraging and allowing everyone to gain a greater understanding of one another."

See also Project Angel Fares below, for possible assistance with transport from within continental US to Morgan's Wonderland.

Read more: [Morgan's Wonderland](#)

## Project Angel Fares

There are children with special needs all across the country and for them amusement parks may be something they never get to truly experience. Project Angel Fares serves to provide all travel expenses paid trips to Morgan's Wonderland, the world's first ultra-accessible theme park, where anyone can play!

Read more: [Project Angel Fares](#)





## 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 the importance for carers of asking for help, and of keeping connected.

### Asking for Help

An important resilience tactic that we can overlook is asking for help. Resilience isn't only about developing our own capacities, it's also about recognising when help would be useful and asking for it, drawing on the skills, knowledge, experience and love of those around us. Science is showing however that there are benefits from seeking help from others, beyond the obvious.

#### **Asking for help, helps us and others too**

Sometimes asking for help isn't easy – as carers we may be too busy or tired to take a step back and recognise that we might benefit from a bit of help or we could be reluctant to ask because of pride ('I don't want people to think I can't cope') or not wanting to impose on others ('I don't want to bother anyone'). Or maybe we're comfortable asking for expert, professional help such as from such as doctors, but not for more day-to-day things for ourselves.

But perhaps we should think differently. Recent scientific research is showing that helping others is an important source of wellbeing – not just for the recipient of that help but for the helper too! So we should think of asking for help not as a burden on others but as an opportunity to boost their wellbeing! It shows that you value what they can do and it can help build your relationship, which is good all 'round!

#### **What help would be helpful for you?**

Leaving aside situations that need specialist help, what help would be most useful? Are there things that you find challenging or hard to fit in? It could be for small things like filling in forms, getting the shopping or tidying the house. Or someone to step in for an hour to allow you to take a short break. Or maybe you want to learn a new skill, find ideas for different ways of doing things or track down information.



Don't wait until you are at breaking point, it's resilient to proactively ask! Who do you know who might be able to help? It might be another family member, a neighbour or friend. What do they really enjoy doing? How might this be helpful for what you need? Patient associations and other carers are also good sources of information, ideas and resources.

The [guide attached](#) has some further questions that you might want to reflect on.

Importantly don't forget to show your appreciation. A simple thank you goes a long way for those that help us. Be sure too, to say specifically what it was that was helpful and the positive impact it had for you.



## Keeping Connected

Human beings are social creatures; we thrive on a sense of belonging and companionship. Research shows being and feeling connected to others is vitally important for our wellbeing and resilience. Both our 'loose' connections (to people who we see regularly but aren't close to) and our 'tighter' connections (those nearest and dearest to us) matter.

### **Our loose connections – mini moments matter**

People who know their neighbors (and say hello to) feel safer and happier where they live. Likewise at work or in groups we belong to, being seen, considered and included feels good whereas the opposite doesn't. So the few seconds it takes to smile and say 'hello' or to ask how they are aren't just nice things to do, these actions make a big difference to our own wellbeing and to others too.

### **Nurturing our close ties**

Our close relationships are the most important for our wellbeing. Yet it is often these that we take most for granted. As busy carers, our attention and energy is mostly focused on the person we are caring for, and it can mean that we have less time and energy for other people who are important to us. So what are some simple and quick ways we can nurture our other close relationships?

A good place to start is bringing to mind what we really love and value about the other person. The best qualities we see in them. Research suggests that if we spend a little time focusing on what's best about them before we spend time with them, that time is more enjoyable and more impactful all round.

In our very close relationships we can often focus on what others do that irritates or "niggle" us, which means we notice more of that! This can especially be the case if we are tired or anxious. Focusing on the niggles impacts our interactions with the other person and ultimately the quality of the relationship.

So who is someone you know you are going to spend some time with today or tomorrow? Before seeing them, spend a few moments thinking about what's best about them. Then, see what you notice about how you feel when you are with them.

### Listening and responding well

The way we listen and respond to others is also really important for the quality of our close relationships. Not just when others need our support but when they have something good they want to share. We can respond in different ways but not all of these nurture our relationships, in fact some are actually destructive. Check out the [guide attached](#) to see how you generally respond to those close to you and try out a way that builds our connections to those that matter most to us.



Thanks to Judy Bell-Broersma for this meme



## ISMRD warmly welcomes to our family



- Malynda Norstadt, who is 40, has ML III and lives in California
- Ana Karin Garcia Alvarez, who lives in Mexico and whose son has Mucopolysaccharidosis or Gangliosidosis
- Holly Thompson whose daughter Madison has ML II/III
- Deborah Feagle and Michael Sounart who care for Rebecca Feagle – Alpha Mannosidosis
- Dana and Travis Faerdy whose son Quin has Alpha Mannosidosis
- Mahendra Patel whose child Tamosjq has Sialidosis
- Silvia Blance whose son Hugo has ML II
- Deanna Barny whose child has ML III
- Shirley Jamil whose son Samuel has ML III
- Donatas Mikuta and Gintare Mikutiene whose child has Alpha Mannosidosis
- David Sowers and Charlotte Gilbert whose son Hunter has Mucopolysaccharidosis III



**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](mailto:info@ismrd.org)**

ISMRD is happy to announce that there have been no bereavements among our member families that we are aware of since the last newsletter



**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**

- Sarah Noble, ML III, New Zealand
- Hayden Noble, 33, ML III, New Zealand
- Karin Blomberg, Aspartylglucosaminuria, USA
- Andre Andrews, ML II, 21, Washington DC
- Savahanna Amour, ML III, 28, Arizona USA



## 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

-  Patricia Adkins
-  Christine Austin
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