



A publication of ISMRD



ISMRD attends the World Symposium see Page 9

HIGHLIGHTS IN THIS EDITION	
From the President's Desk	Page 2
Appointments to Professional Advisory Board and Board of Directors	Page 5
From the Fundraising Team	Page 6
Announcement of Family meeting and Natural History Study	Page 8
A Family report from the WORLD symposium	Page 9
What's in the News	Page 14
Family stories	Page 12&16

**The International Advocates for Glycoprotein Storage Diseases**

3921 Country Club Drive  
Lakewood, CA 90712

[info@ismrd.org](mailto:info@ismrd.org)  
[www.ismrd.org](http://www.ismrd.org)

501 (c) 3 nonprofit organization  
FEIN #52-2164838

**Our Mission:**

ISMRD is the leading advocate for families worldwide affected by a Glycoprotein & Related Storage Disease. Through partnerships built with medicine, science and industry, we seek to detect and cure these diseases, and to enable a network of support and information.



# From the President's Desk

*By John Forman  
President ISMRD*



## **Hello to the ISMRD network.**

2010 was a very busy year for ISMRD and I want to acknowledge the many hours of work done by the board to ensure that all our systems were operating efficiently, so that we can move the mission of ISMRD forward.

ISMRD continues to receive direct support from:

- Mark Stark, who covers the cost of accounting fees.
- Lysosomal Diseases New Zealand, who cover some of our office expenses.
- New Zealand Organisation for Rare Disorders, who manage our website content and cover website hosting costs.
- Andrea Gates and Pam Tobey, who help with postage and copying costs in the USA.

Again last year there were some wonderful fundraisers happening around the United States. The Anna James Fundraiser raised \$768. Pam Tobey's fundraisers raised \$2,451, Ethan Finnie Fundraiser raised \$1,250 and the Rock4Dakotah \$5,000. It's really exciting to see these kinds of fundraisers happening out in your communities. These efforts help to raise awareness and support of these very rare conditions. Well done everyone.

Our grateful thanks also go to the Kimmert family who donated \$1,310, Paul Murphy \$500 and Christina Duthie, \$750. We also received a grant from Emil Kakkis of \$10,000 to put towards our family conference, and although this did not take place in 2010 we are able to redirect this to our next meeting in 2012. There were a number of smaller donations totaling \$842. The total raised by families via fundraisers and donations was \$22,871. I want to thank everyone for the support you all give to ISMRD.

Jenny Noble and Pam Tobey spent a good part of last year preparing and submitting 45 grant applications and although this time round we have not been successful with any of these grants, there are a number of grants where ISMRD has been asked to re-apply. There is a huge amount of work that goes into preparing grants and I want to thank Pam and Jenny for their efforts during 2010.

The fundraising committee is investigating other ways to raise additional funds so that we can support research for our diseases, host the 2012 family meeting in the USA and hold a joint scientific and family meeting in Europe (date still to be decided).

Our database continues to grow and although the growth in numbers was small last year we now have 177 families known to ISMRD. Our new web address is working really well, which has seen quite a few families looking for support and information especially for those contemplating transplants and dealing with difficult symptoms.

We are working on improving our communications with families. We only managed to get two newsletters out last year and with our workload for 2011 we expect a similar number of published newsletters for 2011. Some of the board keep in touch with families via the I-Cell facebook page which was set up by families; this is a really active group. The use of the penguin cafe has declined as other social media has increased throughout society. However, it is still a very important tool for ISMRD to have. Also, in keeping with the facebook trend, ISMRD now has a facebook page of its own and we expect that the range of communication systems we have will enable us to keep in touch with all our families.

The new ISMRD banner and brochures had their first viewing at the International MPS and related diseases symposium in Adelaide June 2010. There was an excellent response from the symposium delegates which saw us give out over 200 brochures.

We continue to build our networks around the world. ISMRD was represented at the National MPS Society conference by Andrea Gates and Jane Andrews. We have re-established contact with the Hide and Seek Foundation and we continue to work with Dr Cathey on the Natural History Study.

Early this year the ISMRD board of directors attended the WORLD Lysosomal meeting. This was an incredible opportunity for us, and having so many people from ISMRD on the ground allowed us to network with a good number of people. We also hosted a booth for ISMRD, displaying our banner and handing out many brochures. I want to thank Pam and Carolyn who did a wonderful job raising in excess of \$300 from the sale of jewelry and a raffle all being done from our booth. This was an excellent publicity drive for ISMRD. Well done everyone for taking up this challenge.

After the WORLD meeting ISMRD held its Annual Board meeting as part of a day-long planning session. We also hosted our Professional Advisory board for a 2 hour meeting and then dinner to discuss the future goals and priorities for ISMRD.

With their suggestion and blessings we agreed to invite Dr Sara Cathey, Dr Richard Steet and Dr Thomas Braulke to become additional members of the Advisory board. We are pleased to announce that they have all accepted this role. Including these additional professionals gives a very strong Advisory board across many of our diseases.

During our Annual board meeting we appointed Susan Kester and Jackie James to the Board of Directors. I want to warmly welcome them to the board and I know Jenny, Andrea, and Pam already have them working hard contacting families, looking at fundraising projects for the 2011 year, and assisting with newsletter research. Welcome to the team.

It was wonderful to have Dr Sara Cathey join us for part of our board meeting. Sara spent time bringing us all up to speed on the Natural History study and other research initiatives, and outlined how important our role was in getting families to her for the study. With this in mind, Sara suggested that we hold our next ISMRD Family meeting in Charleston, South Carolina and hold the extension of the Natural History study at the same time. We think this is an excellent idea and are now working towards holding our family meeting on the weekend of 28<sup>th</sup> and 29<sup>th</sup> July 2012, with Natural History Clinics on Friday 27<sup>th</sup> and Monday 30<sup>th</sup> July 2012.

An exciting new step in the work of the European research consortium for Alpha Mannosidosis has seen the phase 2 clinical trials begin for enzyme replacement therapy for this condition right at the end of 2010. We look forward with great anticipation for the results of this trial late in 2011.

2011 is shaping up to again be another very busy year as we work towards raising funds to support the operations of ISMRD but more importantly working towards hosting our family meeting in South Carolina in 2012. We also intend to put more effort into direct funding of research projects for our diseases and will report more about that soon.

With my very best wishes to all of you,

**John Forman**  
**President**

---



## **A Message from Heaven**

*I am not so far from you  
Just a little way beyond  
Past the cares and past the pain  
Far past my earthly bonds  
When you feel you miss me most  
As years go drifting by  
Each memory will prove to you  
That our love will never die  
For our memories are but a touch  
From the fathers gentle hand  
To heal your pain and mend your  
hearts  
To help you understand  
That while I left you far too soon  
I did not leave alone  
For the father sent his angels  
To gently take me home  
Take comfort when you think of  
me  
Keep my love alive in your heart  
And with the warmth of each memory  
We will never be apart.*

### ***Sadly we mourn the loss of***

**Ethan Finne** who passed away on 4/3/2011

He will be sadly missed by all those who knew him.



## ISMRD appoints new members to Professional Advisory Board and Board of Directors

ISMRD is thrilled to welcome to our Professional Advisory Board.



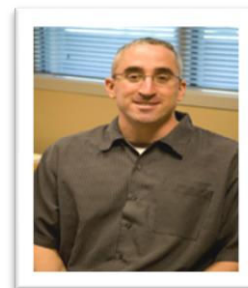
Dr Sara Cathey  
United States



Dr Barbara Burton  
United States



Prof Thomas Braulke  
Germany



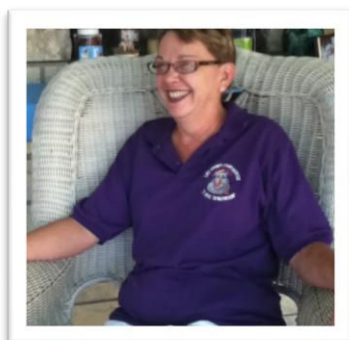
Dr Richard Steet  
United States.

### **Also a part of our Professional Advisory board are:**

Steven Walkley USA, Alessandra d'Azzo, PhD USA, Mark Haskins USA, John Hopwood Australia, Dag Malm Norway, Marc Patterson USA, Charles Vite USA.

---

ISMRD warmly welcomes Susan Kester and Jackie James to our Board of Directors



### **Susan Kester – United States**

Susan Kester is the mother to Lonnie Tice, 23, who is affected by Muclipidosis III. Susan has two other children, Chris and Sarah, who are unaffected. Susan is employed as a cafeteria manager, with the school system in Lee County, Florida. Susan is an active volunteer with the music department at the local high school that all three of her children attend. Susan is looking forward to working with our Glycoprotein Storage Disease families to help accomplish the mission of ISMRD.



### **Jackie James. - United States**

Jackie James is the mother to Anna who is affected by Muclipidosis III. Jackie and her husband Bret also have a son, Peter who is unaffected. Jackie keeps busy running her English tea room in St. Louis, MO, home schooling Anna and taking care of her family. She is looking forward to working toward raising as many funds as possible for ISMRD and helping to move forward the ISMRD mission.



## From the Fundraising Team

*The fundraising team for ISMRD is Jenny Noble, Pam Tobey, Susan Kester, Carolyn Paisley-Dew, Jackie James and Andrea Gates. We are all really excited with the new direction being taken by ISMRD which sees this team hard at work looking at fundraising initiatives with some really exciting things being done by Pam and Jackie (see their articles below). We are actively looking for grants again to support our family meeting in South Carolina and looking for research funding to support future research for our group of diseases. If there is anyone else in our Glycoprotein family considering a fundraiser we would love to hear about it. The fundraising team is ready to jump in and help in any way we can.*

### **Pam Tobey's fundraisers:**

Going to Las Vegas in February and talking to all the researchers and doctors there gave me a whole new spark in my fundraising efforts. I heard, and I see all the potential out there now for ISMRD and all the opportunity that lies ahead of us to make this organization become what we have envisioned for it. I came home and started my planning and telling all my "fundraiser helpers" what goals I had in mind and our planning is really taking off.

We start with a Spring Fling in our town on April 30th and we will participate with a bake sale there. The next two week-ends brings about towns in our area having festivals. I plan on being there to set up booths and sell our jewelry. I guess I need to explain about the jewelry. I have a friend that owns a jewelry co. named Jewel of the Amazon. She is from Bolivia and knows how poor the people are there. She hires the ladies to gather seeds from the Rain Forest and make them into jewelry. It is so unique and beautiful and it really sells itself. (We started in Vegas with outstanding results). She has partnered with ISMRD to sell this jewelry and we get 50% off everything we sell. The jewelry will be offered on the web at a later date.



***An example of the beautiful necklaces that we will have for sale soon via the ISMRD website.***

June 11 we are having a concert with some local bands. In September we have a county fair in our town and we have already reserved a booth there. We have so many things to sell. We will also have clowns coming to help us by making balloon animals and selling them as well, there will face painting and picture taking. We add something new to our list everyday. In May 2012 we are going to do a Tea Party at my church and have activities for little girls, mothers, grandmothers and great grandmothers....if you are female you can come and participate !!!

I would like to challenge each and every ISMRD family to do some sort of fundraiser. It does NOT have to be anything big....whatever you feel that would be appropriate for your family and/or your location. There are so many fundraisers to choose from and if you need help of any kind, I am sure you could just yell and there would be others to help. I could never do what I have done without the help of so many friends and family members. It only took doing the first fundraiser and seeing what we accomplished to make myself and all my helpers so excited to do it over and over again, and don't forget ISMRD now has a big Fundraising team who are only

too willing to jump in and help out where ever they can. Come on ISMRD we can do this it's for our children and loved ones.

### **Jackie James Fundraisers:**

I think I was as surprised as anyone else when I found myself organizing a somewhat successful raffle this past November to benefit ISMRD. It was always someone else other than me that would be an absolute superwoman (or man!) and make the many arrangements to work on such an event and I would quite happily buy a ticket or two. So how did I end up not only engineering a raffle last year, but also end up planning a large benefit in conjunction with the Royal Wedding and a massive dinner/concert event for April 2012?? I believe the moral of this story is that if I can do it, then it is so possible for anyone else to do it too!

In the years that have passed since my daughter Anna's diagnosis, ISMRD has been a doorway for me to keep in touch with other families, meet the most wonderful, caring researchers and doctors, and most of all, provide some semblance of hope that one day there will be a treatment or a cure for Mucopolysaccharidosis. I have to say however, that up to very recently, I never considered that ISMRD needed a considerable amount of financial support to continue and expand its future missions. Of course it makes sense that they would need financial backing to grow, but I quite honestly just didn't give it much thought and believed ISMRD would just somehow keep going!

Once I really thought about it and I realized that ISMRD was not just going to receive money floating down from heaven, I made the decision to work on raising money and awareness to help ISMRD in any way I could. The raffle was actually much easier than I thought it would be. I was pleasantly shocked at how willing my friends (and friends of friends) were willing to help out. I received so many wonderful donations that we ended up with about 20 prizes. We raised \$760. I think it was a great way to test the waters and learn from experience how to step out and begin fundraising. For many of you, it could be something totally different that you try. Anything from selling donuts, pizzas, chocolate etc, organizing a raffle, walk/run, bowling up to a full blown dinner/dance event! My words of wisdom would be "every dollar helps ISMRD". If your kids raise \$15 running a lemonade stand that is \$15 ISMRD didn't have before. Quite literally, every penny helps.



My mother and I own a small British tea room in downtown St. Louis, and on Friday, April 29<sup>th</sup> will be hosting a large event for the royal wedding. We are charging \$40 a ticket for reserved seating, breakfast, scones with Devonshire cream and jam, hot tea, wedding cake, champagne and a souvenir wedding mug to take home with them. We will be selling 40 of these tickets and \$10 of each ticket will be donated to ISMRD as well as 10% of all the days sales. We have secured a large screen with projector and sound provided by a local audio visual store that are donating it all to benefit ISMRD! We are

anticipating that we could get up to a couple of hundred guests throughout the day! We will also have a large royal wedding hamper that will feature all kinds of royal wedding memorabilia that

we will auction off with all the proceeds going to ISMRD. I'm hoping we can raise at least \$1000. This event will be televised live on our local Fox station. I will also be visiting the Fox news station a couple of days before and talking about the event and about ISMRD and mucopolidosis. I am hoping it will bring some much needed attention to our children and ISMRD!

Last but not least, I have booked April 21<sup>st</sup> for a large dinner/dance event here in St. Louis. I have managed to secure a popular local band that are willing to donate their time to us. Now I am searching for a venue that will are willing to donate. I am hoping that we will have up to 200 guests. We will also hold a silent auction. My hope is that we can raise \$10,000.00 for ISMRD with this one event.

I say all this not to make anyone think that it would be impossible, but to say that it is SO possible! For any one of you to step out and do what you can do. It may be \$50, it may be \$50,000!! Whatever it is, I want you to know that every little bit helps out. I honestly would love to see ISMRD being able to provide funding for researchers and a way for families to get together whether it be for support or whether it be for a natural history study. My ultimate hope is that ISMRD will find a way to bring the much desired treatments/cures to our children and all the future children of these glycoprotein disorders.



**ISMRD ANNOUNCES**  
**FAMILY MEETING**  
**AND**  
**NATURAL HISTORY STUDY**  
**CHARLESTON, SOUTH CAROLINA**

**Family Meeting July 28<sup>th</sup> and 29<sup>th</sup> 2012,**

**Natural History Clinics**

**Friday July 27<sup>th</sup> and Monday 30<sup>th</sup> 2012**

***We hope you will join us, block out the dates and start saving your pennies. More information to follow as we start to organise the program and venue.***



## World Symposium 2011

By Colin MacKay

### A FAMILY REPORT FROM LAS VEGAS

Colin, Kathy and David Mackay from Ontario, Canada, recently attended the 7<sup>th</sup> Annual World Symposium presented by the Lysosomal Disease Network and the National Institute of Health (U.S.) held in Las Vegas from February 16<sup>th</sup> to 19<sup>th</sup>, 2011.

This was the first such conference we have ever attended, and we were not sure what to expect. It turned out to be a great opportunity to meet and talk to other families and many health specialists and researchers from all over the world. The contacts we made here will be very useful in the future. We are now more aware of current developments in Enzyme Replacement Therapy and have a better idea about how to deal with governments.

We particularly wanted to finally meet John Forman and Jenny Noble, with whom we have previously corresponded by e-mail. John was the first parent of children with Alpha Mannosidosis that we have ever met and he was able to share some of his great knowledge of this disease and also to show us a presentation on his computer. We also met with the other members of the ISMRD Board (Pam Tobey, Carolyn Paisley-Dew, Mark Stark, and Andrea Gates) and spent many hours meeting and exchanging information with them.

The Professional Advisory Board members of ISMRD were also very generous with their time, talking with us and sharing information regarding Alpha Mannosidosis.

Sara Cathey, Steven Walkley, Marc Patterson, Mark Haskins, and Barbara Burton, were all a pleasure to meet and talk with.

David really enjoyed Las Vegas and talks often about going back. He really enjoyed having meals with everyone as David lives for food!!! He felt quite important attending meetings and being the centre of attention. The Venetian/Palazzo Hotel was also a very interesting experience for David, and the live entertainment in St. Mark's Square was a major attraction for him.

John Forman invited Colin to observe the Annual General Meeting of ISMRD, and this proved to be an interesting experience.

We wish to thank all of the members of the Board of ISMRD for making us feel so welcome in Las Vegas and for taking the time to include us in all meetings.

***It was a pleasure to meet the MacKay family and get to talk with you all. We look forward to meeting you all again soon. Colin has volunteered to be our Canadian contact for ISMRD.***



## ISMRD Board Attends WORLD Symposium

*February 16<sup>th</sup> – 18<sup>th</sup> 2011*

*By Carolyn Paisley-Dew*

The World Symposium was co-presented by the Lysosomal Diseases Network and the (US) National Institute of Health. The goals of the Symposium were to:

- Identify the key diagnostic features of lysosomal storage diseases;
- Describe recent diagnostic tests;
- Describe the relative advantages and disadvantages of each therapy currently available;
- Identify unmet needs of each disease being discussed; and
- Formulate new research ideas for these lysosomal storage diseases.

More than 70 talks were given by specialist speakers from all over the world. Several members of the ISMRD Professional Advisory Board spoke, including Drs Steve Walkley, Marc Patterson and Charles Vite.

ISMRD Board Members attended the World Symposium and were rewarded with considerable enrichment of their knowledge, their networks and their ability to raise awareness about ISMRD and the Glycoproteinoses.

ISMRD had a very fine booth at the Symposium, which attracted a lot of attention to our cause. This attention served to alert us to new grant opportunities and newly diagnosed families. In addition to showing the ISMRD banner and providing leaflets, we raffled a doll's house and sold handmade jewelry from the rainforests of Brazil. These two activities helped to raise awareness of the ISMRD as well as some much-needed funds.



***President John Forman, Dr Steve Walkley and David McKay A. Mann Patient***

The Conference was held in the Palazzo Hotel and, typical of Las Vegas hotels, the Palazzo covered six city blocks, had 8,000 suites and was designed so that you had to pass through the casino to get to your room, to the conference area, to the shops, to the restaurants, to anywhere. Participants were given a map at hotel check-in to help them find the lifts. Several Board Members acquired blisters on their feet from the long walk between the accommodation and the conference rooms, even though they were within the one building. We made several treks to Walgreen's pharmacy to buy bandaids for our feet.

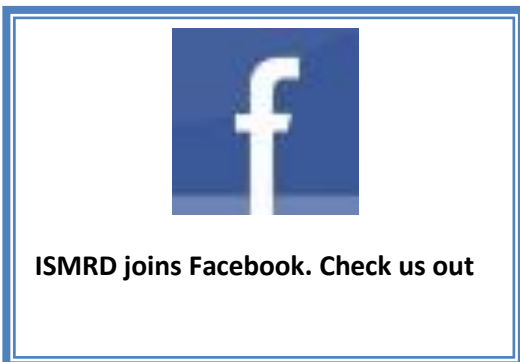
A particular treat for the Board was attending the (US) National Institute for Health's Council of Patient Advocates (COPA), where our opinions were sought on a range of issues relating to patients and families. This was a very valuable opportunity for the ISMRD to have the views of Glycoproteinoses families taken into account in developing policy and deciding direction for research.

There were two days of ISMRD Board meetings following the World Symposium, including a meeting with the ISMRD Professional Advisory Board. Some solid and constructive developments emanated from these meetings, including:

- A date was set for the next ISMRD Family Conference, giving families plenty of time to prepare. After consideration of many factors, the last weekend in July 2012 was decided upon. The ISMRD Family Conference will be held South Carolina, near the Greenwood Medical Centre, so that Dr Sara Cathey can work concurrently with ISMRD family members on furthering the Natural History Study for the Glycoproteinoses;
- The Board developed a three-year plan for research focus and fundraising directions;
- ISMRD Family members Susan Kester from Florida, USA and Jackie James from Missouri, USA joined the Board. Both are enthusiastic about raising funds for furthering our ISMRD goals for research, advocacy and the July 2012 Family Conference;
- It was noted that several family members are making intermittent or regular donations or contributions from fundraising, and that these donations and contributions, however small, really add up to help the ISMRD meet its objectives.



***ISMRD family member Colin Mackay, ISMRD Secretary Jenny Noble and Colin's son David Mackay at the Symposium***



***When in Vegas a wee bit of fun must be had.***

***Carolyn, Pam and Jenny with Will Smith?***



*“How old are your babies?”*

*“Oh wow, they are so small!”*

Those are things we hear a lot around here. I’m the proud mom of four energy filled boys. Ben and Alex, who our 7 year old twins that play every sport under the sun; and Dorian and Wynn, our 2.5 year old twin boys that were born with Mucopolysaccharidosis II and have taught us so much in the 2.5 years they have been here.

After parenting two perfectly healthy boys, when my husband Chris and I found out we were expecting another set of twins we were beyond surprised. We thought *“will another set of twins stretch our time and patience? Will we be able to provide the same amount of loving time with the older boys with two new babies needing our care and attention?”*

We went full steam ahead with this new adventure. The older boys took much pride in helping set up the baby room, putting the old cribs together, and picking out names. They took a lot of pride in knowing that being an older brother came with a lot of responsibilities, and they were excited for the big day of their baby brothers’ birth.

After six weeks in the NICU, the doctors diagnosed the babies with ML2. It was devastating to hear that Dorian and Wynn’s life span would be very limited. That they would only live to maybe 5, maybe 6 years old. How do you tell a parent that their baby comes with an expiration date? Although the news has hit us very hard, we are blessed to have a wonderful team of caring doctors, a supportive family, and an army of loving friends.

I am happy to say that Ben and Alex have taken on the roles of protective big brothers to heart. Alex is always showering Dorian and Wynn with all kinds of love and affection. Ben is the watch dog of the family. If he thinks a nurse or a therapist isn’t doing things right, he’ll be the first to speak up and protect his baby brothers in an instant. It melts our hearts when we watch how much they love their brothers. We have intentionally not told Ben and Alex that the babies have a terminal illness, because we want them to focus on having fun with them *today*, not worrying about what may/may not happen tomorrow.

Dorian and Wynn are only 2.5 years old, but they teach us important life lessons each and every day. Here is just a few of the things they have taught us already:

1. Don’t take a single day for granted. Even if it seems like the day has shaped up to be a “bad” one, there is always room to find blessings in each day and time to turn it into a “Good Day!” as Dori loves to say.
2. God’s grace and love for us is shown in many ways – a hug from a neighbor, a smile on a baby, a good report from a doctor, a new flower blooming in the garden. We have to keep our hearts open to actually see and appreciate these little gifts because it’s the good feelings that comes with those gifts that will be engraved in our hearts forever.

3. Big dreams and little dreams are equally powerful. Before the babies were born we had big dreams for them. Maybe they'd be life-saving doctors, maybe they'd be star athletes, maybe they would be the first twins to be President and Vice President! Who knows?! But we learned that you don't throw away your dreams for your child just because they were born with extreme special needs. You learn to alter your dreams to suit them, and you celebrate each and every little milestone along the way.
4. Don't focus on the negatives. You can't change what has already happened, but you can always change the attitude you use to overcome what has happened. Whether you realize it or not, we are all surrounded by negativity. Negative reports on the news, negative family members, negative co-workers can all form a haze of negativity around you. What you choose to internalize is your choice. Negativity is like food – the more junk food you eat the more it deteriorates your body. Negativity deteriorates the soul, and in our home we choose to keep negativity at the door.
5. Families are amazingly powerful. Dorian and Wynn have helped us create a beautiful family of friends, doctors, a team of therapists (PT, OT, Speech, Auditory), night nurses, sitters, neighbors, friends and loved ones. Family are the people you choose to let into your life, the people you choose to confide in, the people you would do anything for. Dorian and Wynn have strengthened our family beyond anything I could have ever imagined. We are so blessed.

God did not give us more than we can handle. We feel so fortunate that God chose us to be Dorian and Wynn's mommy and daddy. Those little stinkers will continue teaching us the power of love, patience, faith and family. I only hope they can help change your perspective too. Many hugs and blessings from our family to yours.

***The Johnsons,***

***Chris, Mercedes, Ben, Alex, Dorian and Wynn***

## CALLING ALL FAMILIES:

ISMARD needs articles for our next newsletter.

- Family Stories
- Interesting Articles found while you have been searching the web
- Have you been fundraising or planning to do so – tell us about it.





## WHATS IN THE NEWS



### Zymenex enzyme in Phase 2 clinical trials in patients with the rare disease alpha-Mannosidosis

Phase 1 trial has just demonstrated that the enzyme is safe and well tolerated and the Phase 2 dose-finding clinical trial is now underway. The biotechnologically derived human enzyme product rhLAMAN (Lamazym™), which is produced by the Scandinavian biotech company Zymenex and developed for the treatment of patients suffering from the rare disease alpha-Mannosidosis, has successfully completed Phase 1 trials and has now entered Phase 2a clinical trials. The patients were recruited into the Phase 1 trial from around Europe, where the initial goal of demonstrating that the enzyme is safe and well tolerated, has been achieved. This now allows the 10 patients to be moved forward into a 6-month Phase 2a dose-finding clinical trial, where the aim is to identify the most optimal dose to achieve the desired clinical effect.

**Read more at:** <http://www.zymenex.com/PressReleases>



### An Update on Work Carried out by Dr Richard Steet on ML II Zebrafish

Over the last several years, Dr. Steet's laboratory has taken advantage of the speed and utility of the vertebrate model organism zebrafish (*Danio rerio*) to study the molecular and cellular pathogenesis of mucopolidosis II (ML-II). Defining the disease process of ML-II will inform the development of novel therapies that do not rely on the replacement of the defective enzyme. His group's initial findings (*published in the American Journal of Pathology in 2009*) showed that ML-II zebrafish have phenotypes in many of the same organ systems affected in ML-II children. Focusing on the craniofacial cartilage defects, they demonstrated striking changes in the composition and homeostasis of the extracellular matrix in ML-II zebrafish.

These findings are significant since they point to pathogenic mechanisms outside the lysosome and even the cell. More recently, the Steet laboratory has discovered that up-regulation of several classes of protease enzymes also accompany the cartilage defects in ML-II zebrafish. Similar findings were observed in cells from a feline model of ML-II, demonstrating that this up-regulation is a general feature of the disease. His group is now directly testing the contribution of these proteases toward the onset and progression of phenotypes in ML-II zebrafish embryos using small molecule inhibitors and rapid gene suppression techniques.

Preliminary results are encouraging and suggest that a reduction in certain protease activities can partially alleviate the disease symptoms. These experiments will be extended to investigate the impact of the proteases and additional protein targets on other phenotypes within the zebrafish model including

impaired motility and cardiac malformation. This work highlights the power of the zebrafish system to rapidly address disease pathogenesis and test potential therapies.



## Los Angeles Times

### Rare diseases in the spotlight

*April 27, 2011 10:55 a.m.*

Rare diseases are likely to get more attention now that an international consortium of patient advocacy groups and research funders has vowed to deliver 200 new therapies by 2020. For people with these diseases, such attention must seem long overdue.

Drug companies currently don't have much incentive to develop drugs for diseases that affect fewer than 200,000 people, but almost 7,000 rare diseases exist affecting a total of about 25 million Americans.

Many are caused by mutations in a gene. The National Institutes of Health is opening a center in the fall to translate research findings in genetics to usable therapies, the Associated Press reports.

The NIH already has grant programs to spur research in rare diseases. The NIH's Therapeutics for Rare and Neglected Diseases program has a pipeline of projects. Its pilot projects offer a glimpse into some of the diseases that, though rare, can nonetheless have debilitating consequences.

**To read more to go to:** <http://www.latimes.com/health/boostershots/la-heb-rare-diseases-20110427,0,442766.story>

### **The International Rare Diseases Research Consortium (IRDiRC)**

**April 11, 2011**

The diagnosis for most patients affected by rare diseases remains a challenge and most are lacking dedicated therapies. Recent progress in human genomics and other scientific advances increases the prospect for developing effective interventions. Progress will require increased coordination and renewed efforts among multiple stakeholders across the world.

Over the last days a group of research funders, patient advocacy groups, researchers, industry and regulatory agencies continued their work at the second meeting of the International Rare Disease Research Consortium, IRDiRC. The success of this meeting was the endorsement by many stakeholders of the objectives to deliver by 2020 diagnostic tests for most rare diseases and 200 new therapies for patients affected by rare diseases.

This ambitious vision will be realised though an unprecedented cooperation at the international level. The International Rare Disease Research Consortium invites public and private partners with shared commitments from across the globe, to join our efforts and alleviate the suffering of individuals affected by

**To read more go to:** <http://www.eurunion.org/eu/Science-and-Technology/Research/International-Rare-Disease-Research-Consortium-IRDiRC-Statement.html>

## Austin Marine – A Real Hero

**A**ustin turned 13 years old on February 5th. Our young man is now a teenager! When he was diagnosed with ML2 we were told that with the way the disease was progressing he would not live very long. But he is doing fairly well today! I think a lot of his success with maintaining his current state is the nutrition, staying on top of colds and illnesses, and especially keeping up with addressing issues with health such as sleep apnea and the condition of his heart, lungs, bones. It takes going to the doctors a lot, but it has definitely helped him. Austin stands at 32 inches and weighs roughly 35 lbs. He walks, talks, sings, plays the Wii, attends public school, enjoys sports, enjoys attending sporting events with his dad, he has a cell phone and likes to use it, and many other things that 13 year olds enjoy and get into these days. Tonight he is having his best friend from school, Ben, spend the night. There will definitely be lots of ball playing and Wii action this evening!

Physically Austin has had a difficult time with his narrow obstructed airway. He is able to breath on his own with no assistance. His pulmonary and ENT specialists have recommended a tracheotomy, but despite their concerns we have opted to keep him from undergoing that lifestyle. We understand the benefits of a tracheotomy, but feel that Austin's ability to communicate, sing, have sleep over's with friends, and playing in the water at the pool gives him enjoyment and we don't want him to have to give those things up. There things that other children can do that Austin cannot do such as: play on a baseball team with other larger boys his age due to his ability and the danger of getting hurt. It would be very difficult for us to take away some of the things he can do that makes him "Austin". Having said that, he did end up in ICU at UNC Hospital for a week last February due to his oxygen level during a bought with the flu. He recovered fully and after about a month of a raspy voice he is able to speak and sing again like before he caught the flu.

Austin's other major symptom is his bones. A year and a half ago he underwent his third back surgery. His first back surgery was when he was 4 years old. Austin underwent a spinal fusion due to translation (movement) in his spine and the Orthopedic doctor had serious concerns with Austin falling and damaging his spinal column or possible death. Four years later when the translation was still evident with X-rays it was decided to use rods to hold his spine in place. The operations went well, but due to the issue of Austin's spine not fusing from the first surgery and additional surgery was needed to install material to the front of the spine to make the rods fuse in place. The recovery from those two operations was very difficult. We were at UNC hospital for almost 3 weeks in ICU. Austin's most recent surgery was due to translation occurring in his spine above the rods and longer rods were put in to hold his spine in place.

Again, the recovery was the most difficult part of the surgeries. This time it was his airway and oxygen level that kept us in the ICU for over 4 weeks. Each time he was taken off his assisted breathing his oxygen level would plummet. This is when the pulmonary and ENT specialists were recommending a tracheotomy. After we insisted he stay on the machines for a full week before trying to take him off he was able to heal and maintain a healthy oxygen level. We felt he just needed time to let his airway heal after being through everything.

He is a real hero with all he has had to endure. These are just his 2 major physical issues. Like most parents of a child like Austin we regularly visit specialists in many different areas of expertise: Pulmonary, Orthopedic, Eyes, Dental, Genetic, Heart, ENT, Neurology, Cardiology, and Sleep doctor. At night Austin uses a Bi Pap to assist with his apnea and it lets him sleep through the night. Being able to sleep soundly through the night has helped his heart.

Aside from the physical issues Austin has a lot of fun and is not in any pain. He is able to go to public school, sing in chorus, sing at church choir, play the Wii, play in a special soccer league, walk, run (short distances), throw and catch a ball, bat a ball. He has very good eye hand coordination. His small stature and other physical limitations keeps him from doing everything for himself that other children his age can do, but he excels in other areas. He has got to be the most popular child at school and church. I have lost count of the times we have been out as a family eating at a restaurant or shopping and children that we do not even know run up and say that they know Austin. Sometimes it seems his two younger sisters, Courtney and Logan, are a bit jealous of Austin's popularity and attention he gets. There are some people at our church that do not know the rest of our family, but they sure do know Austin. He brightens the room when he is there. Children that are playing rough see Austin and stop what they are doing to toss a ball to him and talk to him because of his personality.

Austin and his dad regularly go to hockey, baseball, basketball, and football games together. They both love the "guys night out". Austin loves to read books and enjoys school. Next year Austin will be in the 6th grade! He is so excited to be moving up to a larger school. His current teachers are sad he is leaving their elementary school and the middle school teachers are already fighting over who gets him in their class.

We feel blessed to have Austin in our family! Austin turned 13 years old on February 5th. Our young man is now a teenager!

### ***The Marine Family***

***Jimmy, Renea, Courtney, Logan, and especially Austin***



ISMRD are the International Advocates for the following disorders: *Alpha Mannosidosis, Aspartylglucosaminuria, Beta Mannosidosis, Fucosidosis, Galactosialidosis, Mucopolipidosis II (I-Cell Disease), Mucopolipidosis III (Pseudo-Hurler Polydystrophy), Schindler Diseases and Sialidosis*



## Contacting ISMRD

### ISMRD Board of Directors

**President:** John Forman | **VP & Treasurer:** Mark Stark

**Secretary:** Jenny Noble

### Directors

United States: Pam Tobey | Andrea Gates |  
Jackie James | Susan Kester

Australia: Carolyn Paisley-Dew

### Founded in March 1999

Postal Details 3921 Country Club Drive,  
Lakewood,  
CA 90712,  
United States

E-mail: [info@ismrd.org](mailto:info@ismrd.org)

Website: [www.ismrd.org](http://www.ismrd.org) | FEIN: 52-2164838

## *Thank you for your Support!*

We wish to acknowledge all these wonderful people who donated funds to ISMRD during 2010:

*John & Susan Bowers, Michael Boyanowski, Angela, Carda, Heather Costabile Thomas & Lisa Crabtree, Mandy Daneman, Freda Derr, David, Dollins, Double Ds Diesel, Drayer Physical Therapy, Penny Dugan, Patricia Dunn, Christina Duthie, E.K. Services Inc. Colleen & Daniel Elder, Andrew Esoldo, Kenneth Faust, Christopher, Finne, James Finne, Tara & Scott Finne, Robert & Caryl Foley, Lawrence Fuller, Janet Fuller, Carol & Arthur Garrett, Kevin & Andrea Gates, Nancy Gautreau Mark Haskins, Heartland home foods, Robert Helgren, Heidi Holmes, Linda & Stephen Henderson, Highmark, Donald & Terri Hilton, Bret & Jackie James, Dick Kailson, Emil Kakkis, Edith Kellermann, Mary Kimmet, Paul & Bettye King, Nancy Kourik, G F Lance, Brenda Lane, Michael & Jennifer Lape, Jo McCullough, Mick Mohriant, Paul Murphy, Jane & Colin Muscroft, David & Tracey Myers, Deborah Oleson, Arthur Ostrander, Brent & Laura Pickett, Earl Radabaugh, Denise, Ramsey, Matthew & Shelly Rhome, Alan & Jodi Richardson, Alan & Patricia Richardson, Richard & Jeraldine Samuelson, Christopher & Amy Schaffer, Sherri & Chaz Scheer, Edward & Carole, Shaw, Thomas & Carolyn Shaw, Snyders Paint Store, William & Carole Spahr, Charles & Patricia Stevenson, Timothy & Janet Stevenson, Heather & William Swartz, Pam & David Tobey, James Trainer, Gregory Troup, Tina Walker, Travis & April Waters, Edward & Nancy Whittmore, Rauol & Gina Williams*



# Feedback Form | Donation

**ISMRD** would like to hear from you! Send us your feedback, your request for further information or make a donation. Just fill out the appropriate boxes below, cut out this page and then return.

Donations: contributions to ISMRD are tax-deductible in many countries. Consult your nation's local or central tax-collection agency. A copy of our current financial statement is available upon request by contacting ISMRD at our address at **3921 Country Club Drive, Lakewood, CA 90712, USA**. Documents and information submitted to the State of Maryland are available from the Office of the Secretary of State or the State Licensing Department. Please contact us for further information.

**Tell us how you can help!** We would like to hear from you and offer you a part in our vision to link families, support research, develop therapies and find cures.

- Send us names and e-mail addresses of family, friends, and professionals who would be interested in receiving our newsletter or who want to know more about our mission.
- Tell us what you can help us with
  - Fundraising
  - Publicity and communication
  - Do you have any other ideas or other ways that you can help ISMRD?

Name: \_\_\_\_\_

E-Mail: \_\_\_\_\_

## Please help our Cause

**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.

**Yes I would like to Contribute the following (check one)**

\$100  
 \$75  
 \$50  
 \$25  
 other\$ \_\_\_\_\_

Please Make Your cheque payable to **ISMRD** Thank you

**Please give us your name & how to contact**

Name: \_\_\_\_\_

Street: \_\_\_\_\_

Street 2: \_\_\_\_\_

City/State/ Province: \_\_\_\_\_

Country/Postal: \_\_\_\_\_

E-mail: \_\_\_\_\_

