Pathways

The Newsletter for Glycoprotein & Related Storage Diseases

A publication of ISMRD

ISMRD announces 2012 International Scientific/Family conference and extension of the Natural History Study

Important dates to mark on your Calendars

31st March 2012 is the final deadline for registering for the conference with ISMRD and booking your accommodation with the Crowne Plaza in Charleston.

See other conference information on page

HIGHLIGHTS IN THIS EDITION

The International Advocates for Glycoprotein Storage Diseases

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

ISMRD

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 Outgoing President's Desk

By John Forman
ISMRD

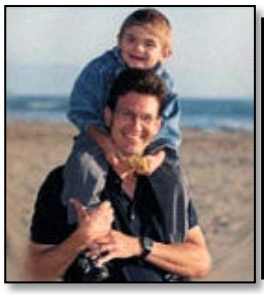
Hello to the ISMRD network.

This is my last editorial as President of ISMRD. I'm stepping down from the role and handing the reins to Mark Stark who has capably filled the role of board member and vice-President for many years now. My time on the board dates back to the beginning of ISMRD in 1999 and my early contact with founding President Paul Murphy. Though it is now several years since Paul stepped down because of work and family commitments, his early work had a lasting impact on ISMRD. It was his vision of an international group for the Glycoprotein storage diseases that made us what we are today and which still guides us into the future.

The financial crisis that ISMRD faced in 2008 led to very challenging times for ISMRD and a lengthy process of rebuilding our activities and communications with families. But we survived that and I'm proud to have played a role along with the rest of the board, in rescuing and restoring the organisation. I have every confidence in Mark Stark's ability to lead ISMRD in the years ahead, though these times will be tough because of the continuation of the global financial crisis that is causing such difficult times for many not-for-profits.

Mark is ably supported on the board by Jenny Noble, Pam Tobey, Andrea Gates, Carolyn Paisley-Dew, Susan Kester and Jackie James, and I want to thank them for the support they have given me over recent years and their dedication to ISMRD. They are now being joined on the board by Melissa Rust and the board promises to be a strong and active one in the years ahead.

Although I am no longer President, I will remain on the board with the role of vice-President for Research. That will enable me to maintain my strong interests in this aspect of ISMRD's activities. In fact I drafted this editorial while on a flight to Paris for a meeting of the Alpha-Mann consortium who are working to complete their research project on Mannosidosis and are part way through a phase 2 clinical trial for the replacement enzyme. So you will hear from me in the future, but with a different slant to my communications.



From the In-coming President's Desk

By Mark Stark
ISMRD

Hello to the ISMRD network.

I am very sad to see John step down as President, although I am very pleased he will continue on our board. I am also extremely honored to be selected as President, and I will be leaning heavily on John and our other fantastic board members. My first experience with ISMRD, like many of you, was when I found the ISMRD web site during a frantic search shortly after getting our son's diagnosis of alpha-mannosidosis. My wife and I were horrified and frightened by this mysterious disease, but we were immensely helped by being able to make contact with other parents of children with rare diseases. First Paul, then John and others whose children had been through what our child was going through were there to talk with us and give us good advice. We had to make very difficult decisions about what to do; and having an experienced, empathetic group of dedicated people to talk to made a huge difference.

I know it is the board's responsibility to make sure ISMRD is always there to support our families. I am very pleased to have been part of the team that weathered some difficult times, and I am very proud that ISMRD was always there with information to help those frightened and bewildered parents who had just gotten terrible news about their child. Of course, we want to do more than just be a source of information and empathy for our families. We aspire to enable early detection of the disease, and to develop effective therapies. We also want to encourage research that ultimately will cure and eliminate these and other genetic disorders. To that end, even though our fundraising has been modest, we have been able to organize and fund our fourth combined scientific and family meeting which will take place this July in Charleston, SC. Once again, experts and researchers from around the world will join together to further their understanding of these diseases and how to treat them. Our families from around the world will have a chance to meet these experts and each other. We know from the previous conferences that new avenues of research will be opened, and life-long friendships will be formed. I hope you can join us there, I am looking forward to meeting all of you.

ISMRD Welcomes New Board Member

Melisa Rust is mum to Meghan who has ML II. She is passionate about being able to help move ISMRD's mission forward and looks forward to communicating and meeting with many of the ISMRD families.

IMPORTANT DEADLINES

FOR

2012 INTERNATIONAL CONFRENCE FOR

GLYCOPROTEIN STORAGE DISEASES

ISMRD is pleased to bring you another family meeting combined with a scientific meeting about the Glycoprotein Storage diseases plus the extension of the Natural History study. Attention and scrutiny will be paid to ***Alpha-Mannosidosis, Aspartylglucosaminuria, Beta-Mannosidosis, Fucosidosis, Galactosialidosis, I-Cell Disease, Pseudo-Hurler Polydystrophy, Schindler Disease and Sialidosis.*** The conference is being held in Dr Sara Cathey's home town of Charleston, South Carolina July 28th – 29th 2012

The conference dates are broken down as follows:

- Scientific conference July 28th -July 29th
- Natural History Clinic days - Friday July 27th & Monday July 30th
- Family conference - July 28th - July 29th

Scholarship Program: We are delighted to have in place a very strong Scholarship program which will ensure that all families have the opportunity to attend the conference. We have broken the scholarship into two parts one for accommodation the other for families who will incur high costs for Air Travel. The details are as follows.

Accommodation Scholarship

For families who book their accommodation with the Crowne Plaza and complete their conference registrations with ISMRD before the **31st March 2012 - *ISMRD will cover the cost of 2 nights' accommodation @ $218+ taxes.*** *This offer is available to the first 60 rooms being booked by families.* We currently can only consider ***one room per family*** but if there is indication that a family might need a second room we will very carefully consider if we can stretch the available funds.

International/Domestic Air Travel Scholarship

Our aim is to give as much assistance as funds allows. This scholarship is intended to assist families who may incur high travel costs. Please contact Jenny Noble jenny.noble@xtra.co.nz for assistance.

Book your accommodation with the Crowne Plaza before 31st March 2012,

Send your registration forms and fees to ISMRD before 31st March 2012,

ISMRD 2012 INTERNATIONAL CONFERENCE

DRAFT FAMILY PROGRAM

Registration
5.00pm – 6.00pm Friday 27th July 2012

Welcome Reception
7.00pm – 8.30pm Friday 27th July 2012

Saturday July 28th		
Joint Scientific/Family Session		**Chair:**
8.30am	Welcome, introductions, opening remarks	John Forman, Sara Cathey
8.45am	Keynote speaker – Overview of glycoproteinoses, old and new questions – Looking to contemporary research for answers	Richard Steet
9.15am	Effective treatment for rare disease – How do we get there	TBA
9.35am	Cerebellar pathology and Ataxia as therapeutic outcome measures for ERT in Alpha Mannosidosis	Judith Blanz
10.00 – 10.30	**Morning Break**	

	Scientific Program	**Chair:**		**Family Program**	**Chair:**
10.30	Translating science to therapy for galactosialidosis	Sandra D'Azzo	10.30	Glycoproteinoses – Highlighting the challenges	
10.50	Haematopoietic stem cell transplant in glycoproteinoses	Troy Lund	10.50	Genotype phenotype correlation in the glycoproteinoses – Mucolipidosis as an example	TBA
11.10	Animal model of glycoproteinoses found in nature and in laboratories	Mark Haskins	11.10	Overview of therapeutic approaches	TBA
11.30	Platform talk(s) from submitted abstracts	TBA	11.30	Understanding neurologic symptoms and signs	Marc Patterson
			11.50	Questions and answers	

12.15 – 1.15	**Lunch and poster viewing**				
	Scientific Program	**Chair:**		**Family Program**	**Chair**
1.15	Bone disease – Before, during and after	Jules	1.30	Bone marrow transplant in alpha	Troy Lund

	dysostosis multiplex	Leroy		mannosidosis	
1.35	Crossing the Blood/Brain Barrier	Bill Sly	1.50	Family experience of BMT	TBA
1.55	The schindler Enzyme Behaves differently	Tim Wood	2.10	Managing skeletal complications in glycoproteinoses (hips, shoulders, spine, hands, feet)	John Davids
2.15	Excessive Activity of Cathespin K and cartilage defects in a Zebera fish model in Mucolipidosis type II	Richard Steet	2.30	Pain management in glycoproteinosis	TBA
2.35	Sialidosis - (Junior Investigator from Dr. d'Azzo's lab	TBA			
	Questions and answers		2.45	Questions and answers	
3.00 – 3.30	**Afternoon Break**				
	Scientific Program	**Chair:**		**Family Program**	**Chair:**
3.30	Platform talk(s) from submitted abstracts		3.30	Ophthalmic complications in glycoproteinoses	TBA
4.00	Questions and Answers		3.50	Dental care in Glycoproteinoses	
			4.10	Family experience of managing dental care for their child	Nila Coley
4.30 – 5pm	Poster viewing sessions		4.30	Poster Viewing session	
7pm	**ISMRD Gala Dinner**				
	Sunday 29th July				
	Scientific Program	**Chair:**		**Family Program**	**Chair:**
8.30	Lysosomal trafficking and exocytosis	Andrea Ballabio	8.30	Managing ataxia and seizures – What to do	
8.50	Proteomics of the lysosome	Peter Lobel	8.50	Getting the right clinical care for patients with glycoproteinoses	John Forman
9.10	Tandem mass spectrometry and screening for oligosaccharidoses		9.10	Natural history study – What are they and why are they important – Update of study	TBA
9.30	Platform talks from submitted abstracts		9.30	Questions and Answers	
	Joint Scientific/Family Session				**Chair:**
10.30	TBA				

10.50	TBA	
11.10	TBA	
11.30	TBA	
11.50	General discussion	TBA
12.15	ISMRD's future direction for research and fundraising	TBA
12.45	Closing remarks	TBA
1.00	Shared Scientific/Family Lunch	

This program is a draft and an indication of what is planned for presentation. Keep watching the website for program updates.

We warmly welcome to our Penguin Family

- Glen Nicholas and Julia Sutton who live in Australia and have a daughter Maddie who has ML III
- Ahsley Pauls who lives in USA and has a son with Galactosialidosis
- Yasar Suleymanbey mah who lives in Turkey and has a child with Alpha-Mannosidosis
- David Vidal who lives in Spain whose brother and sister have Aspartylglucosaminuria
- Jesse and Liz Sibert who live in the USA and have a daughter Mary who has MLII/III
- Julie P Dennison who lives in USA, and whose son 9 year old son Cameron has Fucosidosis
- A Spanish-speaking family in USA whose 3 year old boy has Fucosidosis (if anyone speaks Spanish and would be willing to talk to this family, please email info@ismrd.org)

LIFE WITH DISABILITY

By Sylvia Webb
Australia

Sylvia and family in 1999

Sylvia Webb is an Australian parent with five children born with ML II/III and two without. She is a Christian and lives in Melbourne, Victoria with her husband Charles and her daughters Tegan and Grace. This article is the first in a series of subjects Sylvia has presented to Charles Sturt University Speech Pathology students over the past seven years to help them understand what it is like to be the parent of a child with a disability. Other subjects will be reproduced in forthcoming ISMRD newsletters.

In this article, Sylvia presents an allegory for how we and the general public can inadvertently form misguided views about life with a disability.

THE SALESMAN

A salesman had to travel to an important appointment in a town he had never been before. His destination was a long way from home and so he left early. After several hours he came to a town about half way to his destination.

The town the salesman arrived at was not very large but the town had been designed in a way that all the buildings and homes were centered along the main street. So, as the town grew the main street had grown along with it. And, to pass through the town meant you had to travel across the main street and the highway on which the salesman was travelling was the only way through.

On the day the salesman arrived the town was celebrating a very important event to the town and everyone wanted to attend. Weeks and months had been spent planning the parade and other activities that would be taking place during the event with everyone doing their part to make it a memorable day. It was basically an all-day event when the town was practically closed down.

On seeing the road through the town blocked the salesman entered a service station to get petrol and directions around the town. His appointment was important to him and he just wanted to get to it as soon as possible.

The man attending the service station was a recluse who had experienced many sad and desperate times. Over time he had developed tics and twitches in both his speech and his body which increased in times of stress and meeting strangers was something he found very stressful.

The salesman approached the attendant and asked him for direction to get around the town so that he could get to his appointment, stressing the importance of his appointment. This put the attendant in an even more stressed state and as he started to explain to the salesman the way to get around the town he began grunting and twitching. The salesman had to pay increasing attention to understand what the attendant was saying and was increasingly distracted by the twitching and grunting.

After a while the salesman was happy with the directions he had been given and set off.

He drove along the perimeter of the town into a farming area where the road meandered around the countryside for what seemed like hours. Finally he found himself travelling along the other side of the town and when he saw another service centre he pulled in to get directions to get on his way.

The man attending this service centre was also a recluse. He had dabbled in some illegal substances in his youth which had left him with slurred speech and a mind that wandered. This was fine with those who knew him but difficult for those who didn't. As he slowly started to give his directions to the salesman his mind began to wander to the point that salesman had to continually draw him back to the here and now to find out how to get out of the town he was in and to the town he wanted to go. Eventually he had his directions and set off.

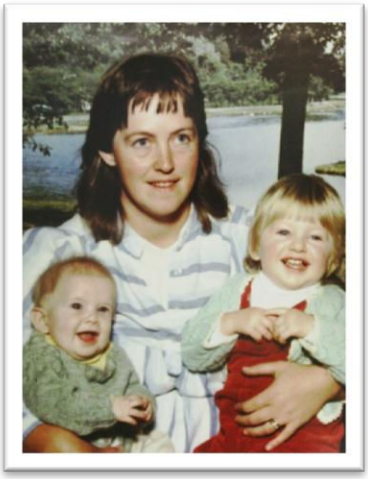
As the salesman drove to his destination he pondered on the type of people who lived in the town he had just passed around and he considered the two men he had encountered and wondered if they could be examples of what the rest of the population were like.

And so it is with us and having a child with a disability.

As we pass through life we more often see or hear about the extreme cases of children and adults with disabilities – the gross, the disfigured, the mutilated, the destructive and self-destructive, the hyperactive, etc. etc. And then, when we are told we have a child with a disability we think of all the stories we have heard and all the badly-behaved children we have seen and wonder if that will be what our child will be like and the prospect is daunting and we become filled with fear and loathing for our future.

The salesman only met two men of the entire population but his mind told him that the rest of the town was just like them.

Some Years ago in 1985 I was given a tape with this story on it. On the tape the man speaking was talking about the extremes we often see or experience in Christians. I can't remember his name. I wish I did, because I'd gladly give him credit for impacting my life with his story not only in the area of being a Christian but also in the area of having a disabled child.



When our daughters Jade and Tegan were diagnosed in 1984, it was one of the most devastating events of our lives. The diagnosis and prognosis given to us was so painful it has hard for us to believe. We wondered what our lives and our children's lives would be like and like so many people we thought the worst. We made a decision that seeing as our children would not have much quantity of years to their lives, we would add as much quality to their lives as we could and fill it with opportunities to live well and reach their maximum potential and we were fortunate to be able to do it.

Sylvia with Tegan (6mths) and Jade (2yr) just before they were diagnosed in 1984

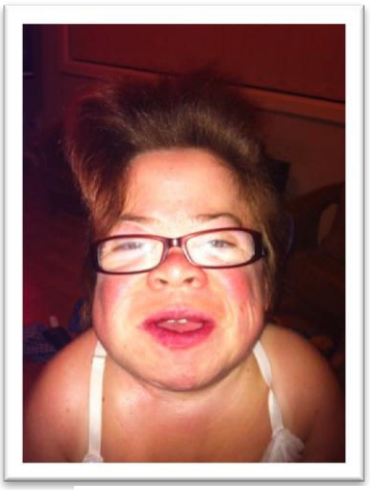
Eventually we decided to have more children. Twins Caryl and Lucas were born in 1985, Christian was born in 1987, Faith in 1996 and Grace in 1997. Of our seven children, Jade, Tegan, Lucas, Faith and Grace were born affected by ML II/III. Jade passed away in 2003, aged 21. Lucas died in 2009 aged 23 and Faith died on Valentine's Day 2010 aged 14.

Lucas "a boy from the bush", 2004

During our lives since we started our family we have experienced many happy and sad, carefree and painful events throughout their lives and we continue to do so with those still with us (Tegan, Caryl and her two sons, Liam and Chase; Christian and Grace).

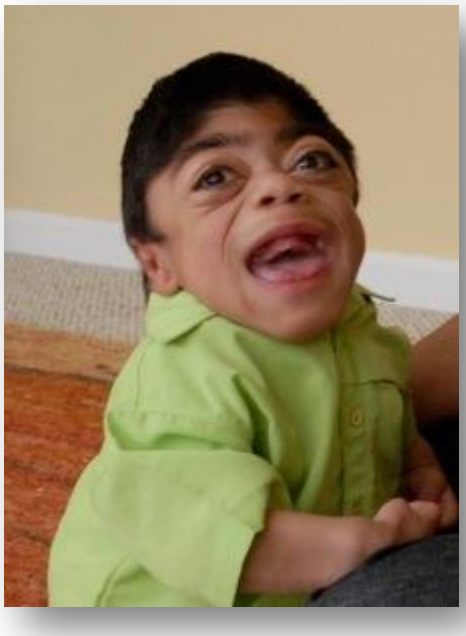
We try not to make judgements (even though it is often hard) about our children or our circumstances by what we've experienced in the past or what we've heard or what we've seen. Obviously we have learned a great deal. Our time with the children is never long enough for any of us but we try to make our time with them time filled with love.

Grace and Faith, July 2009



Tegan 2012

Yash Gandhi Foundation Awards $20,000 to Greenwood Genetic Centre for Read through therapy research for ML II



The Yash Gandhi Foundation has awarded a $20,000 research grant to advance research in I-cell disease (also known as mucolipidosis II or ML II) to Greenwood Genetic Center. This grant was donated in memory of Yash Gandhi who passed away in November 2009.

Dr. Sara Cathey, the Primary Investigator, will be researching the effectiveness of the compound PTC124 (Ataluren) in reading through nonsense mutations in fibroblast skin cultures obtained from children with mucolipidosis II (MLII). In this study, skin fibroblast cultures from patients with MLII will be treated with PTC124 in three different doses to determine if the compound will be effective in partially restoring the reduced phosphotransferase activity.

ISMRD would like to thank the Yash Gandhi Foundation for this ML II research grant. The Foundation is located in Pennsylvia, USA, and can be contacted at asheshg@mac.com.

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http://www.goodsearch.com/about.aspx#faq70

AUSTRALIAN ISMRD MEMBER RAISES AWARENESS

Search for answers can lead to despair

From the Campbelltown–Macarthur (NSW) Advertiser community newspaper
BY ALEXANDRA PLEFFER

Raising awareness: Allison Dennis, 29, from Macquarie Fields has mucolipidosis 3 and hopes to shed some light on the challenges facing people with rare conditions for Rare Disease Day today.

WHEN Allison Dennis was diagnosed with a rare disease at the age of six, her mum didn't know what the disease was or anyone whose child had it.

Devastated, Trish Dennis searched for hours through hospital libraries and asked doctors for any information about mucolipidosis 3. The Macquarie Fields woman said it took a while for her daughter to be diagnosed because such a rare condition didn't occur to the doctors.

"I was devastated," she told the Advertiser. "It destroys you because when you have a child you have hopes, dreams, and ambitions for that child and then all of a sudden everything is gone."
Mrs. Dennis said the internet and Facebook now provided support and information about rare diseases, but a lack of awareness and funding was a challenge.

Today is Rare Disease Day, and she hoped residents would take a minute to think about the struggles facing people like her daughter who live with a rare disease or condition. "I always think that people are scared of what they don't understand," she said. "Quite often, my daughter is in a wheelchair and people will pull their kids away from her in the shops.

"It's about awareness in the community of so many rare diseases that don't have funding and therefore don't have treatments and cures." Mucolipidosis 3 is a genetic degenerative condition with no treatment and no cure. It causes enzymes in lysosomes to leak, continuously damaging the body.

We would love to know what you did for International Rare Disease Day. Please send your story to info@ismrd.org

World Report on Disability

From Rare Gems Newsletter Dec 2011

The inequities faced by people with disabilities in everyday life are well recognised in the global arena. In 2008, the first comprehensive human rights treaty of the 21st century (the Convention on the Rights of Persons with Disabilities, CRPD) came into force.

And now there's been another major step forward. In June 2011, the World Health Organisation and the World Bank jointly produced the first ever World Report on Disability.

In the Report's Preface, Dr Margaret Chan (Director-General of WHO) and Mr Robert B Zoellick (President of the World Bank Group) state:

"Our driving vision is of an inclusive world in which we are all able to live a life of health, comfort, and dignity."

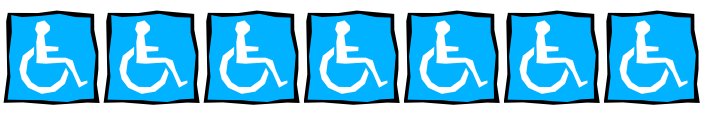
The definition of disability has been extended, including those who suffer from any decreased level of functioning, meaning that conditions such as diabetes and cardio/respiratory problems qualify under the new definition.

The Report identifies barriers, suggests recommendations for change, with responsibility for such change placed on everyone.

Despite examples of good practice within the Report, the expert authors do not turn a blind eye to the current global disability scene. One of these authors, Dr Tom Shakespeare, was quoted in The Guardian as saying: "The clear message from the report is that there is no country that has got it right. Italy is a world leader in terms of inclusive education and de-institutionalisation of people with mental health problems but in other areas it is not. In the US the access is phenomenal - it is a civil rights issue. However, if you are looking at poverty and employment it is not good."

For more information go to: http://www.who.int/disabilities/world_report/2011/en/index.html to read the **full Report, Summary, and a fact sheet.**

And http://www.who.int/mediacentre/multimedia/podcasts/2011/disability_20110610/en/index.html to here to hear an **enlightening podcast, hosted by Dr Tom Shakespeare, one of the authors and editors of the Report**



TRANSFORMING THE REGULATORY ENVIRONMENT TO ACCELERATE ACCESS TO TREATMENTS (TREAT ACT)

from US National Organization for Rare Disorders (NORD) newsletter

On 15 February 2012, Senator Kay Hagan introduced the Transforming the Regulatory Environment to Accelerate Access to Treatments (TREAT) Act. The following is the text of the statement made by Hagan. It mentions the need to ensure uniformity in the application of flexibility.

The proposed legislation:

- enhances and codifies the accelerated approval process;
- addresses concerns of the rare disease community related to conflict of interest provisions;
- provides greater clarity, consistency, and transparency in review processes; and
- encourages innovation and adoption of modern scientific tools in regulatory science.

To read more go to http://www.hagan.senate.gov/files/TREAT_Act.pdf

SENATOR HAGAN'S STATEMENT

S. 2113. A bill to empower the Food and Drug Administration to ensure a clear and effective pathway that will encourage innovative products to benefit patients and improve public health; to the Committee on Health, Education, Labour, and Pensions.

Mr. President, today I am proud to introduce the Transforming the Regulatory Environment to Accelerate Access to Treatments (TREAT) Act.

This bill empowers the Food and Drug Administration to ensure consistent processes and a clear and effective pathway that will encourage the development of innovative treatments to benefit patients, particularly subpopulations and those with rare diseases, and improve the public health.

Without question, the FDA plays a critical role in helping to ensure that new medicines are safe and effective. At the same time, by promoting investment in and development of innovative treatments for unmet medical needs, the FDA can positively influence our national strategy to identify and treat serious and life-threatening diseases and improve the quality of life for millions of Americans.

In order for FDA to accomplish this goal, however, Congress needs to give the agency the tools necessary to transcend existing barriers, reform its processes, and provide greater clarity, consistency, and transparency to industry.

The bill accomplishes this in three ways.

First, it provides the FDA with the authorities and tools that are reflective of the agency's responsibilities and that are necessary to ensure maximum operational excellence by updating FDA's mission statement and creating a management review board.

Second, it advances regulatory science and innovation within FDA to ensure that evaluations of innovative treatments, therapies, and diagnostics are conducted by those who have the best available knowledge. To do this, the bill creates a chief innovation officer and chief medical policy officers, and expands participation on advisory committees by those experts most familiar with the disease being considered.
Finally, the bill promotes the utilization of modern scientific tools and methodologies to ensure patients have timely access to innovative products by creating a clinical informatics coordinator, providing more information to drug sponsors when an application has not been approved, and enhancing and codifying the accelerated approval process.

In the nearly 2 decades since the accelerated approval mechanism was established by FDA to more expeditiously approve treatments, advances in medical sciences, including genomics, molecular biology, and bioinformatics, have provided scientists with an unprecedented understanding of the underlying biological mechanisms and pathogenesis of disease.

A new generation of modern, targeted, personalized medicines is currently under development to treat serious and life-threatening diseases. Some apply drug development strategies based on biomarkers or pharmacogenomics, predictive toxicology, clinical trial enrichment techniques, and novel clinical trial designs, such as adaptive clinical trials that can be altered based on observed patient outcomes in the interim.

In order to ensure these scientific advances are translated into treatments that benefit patients, Congress should allow FDA to implement a more effective process for the expedited development and review of innovative new medicines intended to address unmet medical needs for serious or life-threatening diseases or conditions.

FDA is already doing this, to some extent. However, application of the accelerated approval process has been somewhat limited, largely to HIV and oncology drugs, and inconsistently applied to other disease targets. For example, a 2011 report by the National Organization for Rare Disorders compared the approval process for 135 non-cancer orphan therapies approved by FDA from 1983 through June 2010. The report found that 45 went through the conventional approval process; 32 were approved with some sort of administrative flexibility; and 58 were approved on a case-by-case flexibility process. This report illustrates that while FDA does have the authority to approve these treatments with some flexibility, there does not appear to be uniformity or consistency in employing this flexibility.

The TREAT Act allows FDA to tap into modern scientific advances by using a broad range of surrogate or clinical endpoints and modern scientific tools earlier in the drug development cycle, when appropriate, to approve treatments for patients. Employing these modern scientific tools may result in or shorter clinical trials for the intended patient population or targeted subpopulation without compromising or altering FDA's existing high standards for the approval of drugs.

It is the patients suffering from these serious and life-threatening diseases that benefit from expedited access to safe and effective innovative therapies. For the 30 million Americans living with rare diseases, new advances in science and medicine cannot come fast enough. That is why I am proud that this bill has the support of the National Organization for Rare Disorders (NORD) and Friends of Cancer Research. The TREAT Act provides FDA with the tools needed to modernize its processes and encourage the development of innovative products to benefit patients, particularly subpopulations and those with rare diseases.

I urge my other colleagues to join us in supporting this important bill.

THANK YOU SENATOR HAGAN, FROM ALL AT ISMRD

Some of our Penguin children and young adults have had surgery or are on the waiting list for surgery.

Your Penguin family are thinking of you all and praying for good outcomes and speedy recoveries.

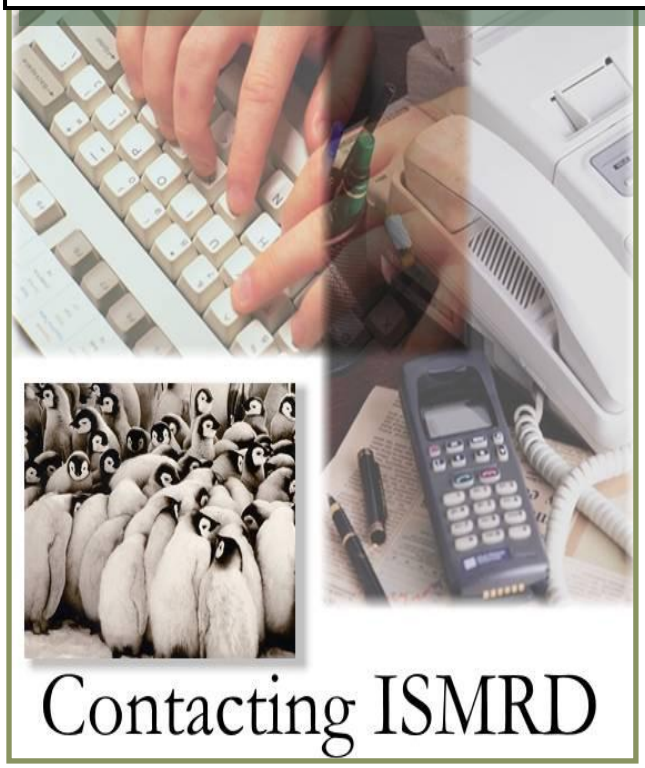
- Meghan Rust who has had surgery and is recovering well.
- Autumn Tobey has had a shoulder replacement and is recovering well
- Tessa Nelson is waiting for spinal surgery on 21st May
- Sarah Noble is waiting for Shoulder replacement.
- Alie Dennis is to have investigations due to shoulder pain and is having another spinal operation on 22nd March.

ISMRD Honor Roll 2011

Thank you to everyone who provided funds for the ISMRD in 2011 by donating, fundraising or grant writing

Marion Abdullah	*Genzyme*	*Pam Lyon*
Blair & Carolyn Anderson	*Global Impact Applied Materials*	*Teresa & Sam Montoya*
Kimberly Bedell	*Good Search*	*Angela Mulligan*
Donald Carlos	*Highmark Blue Shield*	*Jenny Noble*
Gustavo & Maria Elena Cárdenas	*Ken & Kathy Hirabayashi*	*Vincent O'Connell*
Frank Coffey & Cris Lopez-Coffey	*Philip & Carolyn Holzman*	*Carolyn Paisley-Dew*
Dan Bindman & Norma Edwards	*Scott & Lynn Hopkins*	*Judy Rompf*
Dennis & Dianna Bisnar	*William & Joyce Ingram*	*Scott & Greta Salhus*
Michael & Kathryn Brown	*Bret & Jackie James*	*William Skojec*
Citgo	*Susan Kester*	*Dana Shumaker*
Thomas & Lisa Crabtree	*Kimmet Family*	*Mark Stark*
Allan Cuilty	*Paul & Bettye King*	*Elie & Tanya Suares*
Mary Anne Cuilty	*Allan & Su Pegg Lane*	*Luis Suarez & Haydee Testamarck*
David Dollins	*LDNZ*	*Allan Thomson*
Double Ds Diesel	*Lehigh Valley Chapter of Music Teachers*	*Pam & David Tobey*
Christina Duthie	*The London Tea Room*	*Richard White*
Lee Anne Ellison	*Megan Luther*	*Peggy Wilson*
Charlie & Cindy Ewers	*Marina Oil Services*	*Woolley family*
Kathleen Gann	*Jaan Mead - Bedevoted*	*Mark Yamamoto*
Gates Family Trust	*Tom & Selma Mirante*	*Mark & Rita Zaarour*
Kevin & Andrea Gates	*NZORD*	*Loyda & José Zapata*

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



ISMRD Board of Directors

President: Mark Stark
Vice President, Administration: Jenny Noble
Vice President, Fundraising: Pam Tobey
Vice President, Research: John Forman

Directors

United States: Jackie James| Andrea Gates | Susan Kester | Melissa Rust
Australia: Carolyn Paisley-Dew

Founded in March 1999

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Lakewood,
CA 90712,
United States

E-mail: info@ismrd.org
Website: www.ismrd.org | FEIN: 52-2164838

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, **please contact Susan Kester**. She will coordinate with the appropriate parties to determine how we can best help.

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

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: _____