



A publication of ISMRD



**ISMRD announces Family meeting**  
**Las Vegas 19-20<sup>th</sup> February 2011**



**ISMRD launches its new**  
**Publicity materials**


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**The International Advocates for Glycoprotein Storage Diseases**

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 Lakewood, CA 90712

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[www.ismrd.org](http://www.ismrd.org)

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



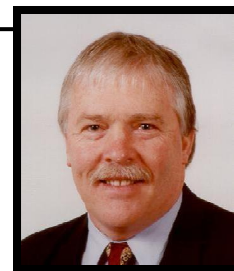
**Our Mission:**

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.

*My message for this news letter is exactly what I reported to the ISMRD board at our recent annual meeting .*

*This is the annual report for ISMRD for the 2009 year.*

2009 was a very challenging year for ISMRD and I want to acknowledge the many hours of work the board has put into restructuring ISMRD's operations. It has been quite a challenge to organise some items across borders, and I'm very pleased with the way the board members have ensured continuity of all our administrative processes, while still managing to look towards future projects.

In this our first Pathways for 2010 you will see reports on some of the projects we have been working on and stories from families who have been raising funds to support the mission of ISMRD.

There have been some fantastic fundraisers organised throughout 2009 and the early part of 2010. The Charity Dinner held in California by the Gates family raised \$32,000. This fundraiser helped stabilise our very precarious financial situation. Other fundraisers held were the Rock for Dakotah, Ethan's Slide Glide and Ride bash and Pam Tobey's music CD sales. These are exciting innovative ways of helping us raise funds. Thank you to all those families who have been involved. We also have financial support that comes to us via other means such as the Kimmet family monthly donations, our accounting fees covered by Mark Stark, LDNZ who are covering postage and printing, and NZORD who host the website and cafe. I also want to acknowledge all those families and friends who have sent in donations to ISMRD via PayPal, JustGive or have sent in cheques. Total funds raised including the Gates family's big fundraiser was \$55,600US during the 2009 year. Without your valuable support ISMRD would struggle move our mission forward.

The board has recently submitted a number of grant applications for various costs and projects and we plan to continue these applications on a regular basis from now on. We hope that with persistence ISMRD will become successful in gaining grants to support the wonderful efforts of our families in raising funds for us.

Our projects during 2009 saw ISMRD supporting ML families to attend the Greenwood Genetic Centre for the second stage of the Natural History Study for ML patients, and in November 2009 Dr Sara Cathey and Dr Lucia Horowitz travelled to Australia and New Zealand to gather samples and Natural Histories from 15 affected family members. We are very appreciative of the US\$7000 grant provided in 2009 by the US MPS Society to support this work, with the rest of the costs coming from the research scholarship won by Jenny Noble in the previous year.

Our last annual report discussed a major grant application to the NIH to support Dr Sara Cathey's work on the natural history study and we are pleased to note that a grant of US\$250,000 was given via the Lysosomal Diseases network. This will support the study over the next 5 years.

Our Website and Penguin cafe have had some makeovers and upgrades, and in March this year they were transferred to a new content management system and hosting arrangements that offer more security and ease of updating. Our next plan for the website is to arrange translation into several languages. This is an important part of the International aspect of ISMRD. We have also printed new brochures and a banner for ISMRD.

We have been updating our database of ISMRD family contacts and have moved it to a system that is easier to manage. During this update we saw our database grow from 150 to 175 families. We still need to follow up on

families who have moved and not updated their details. If this applies to you could you send your new contact details to Jenny Noble or use our [info@ismrd.org](mailto:info@ismrd.org) address. That email address is now the primary contact point for ISMRD.

We continue to work on the issues that are important to our group of diseases. Lobbying on issues related to newborn screening for Lysosomal diseases has produced a commitment from New Zealand's screening advisory committee to review screening criteria. A number of US states have begun screening for a small number of other Lysosomal diseases. Both these developments have indirect benefits for us by keeping up the momentum and debate about new conditions to screen for, and we continue to keep a close eye on these developments, working towards the day when our conditions are also screened for in new born screening programs.

Our networks with other Lysosomal research groups continue to be strong. Late last year ISMRD became a partner with the EveryLife foundation set up by Dr Emil Kakkis, to look at how the research and fast tracking of orphan drugs can be improved for rare and neglected diseases. We are also partners in the Global Genes project and you can read more about this in this edition of Pathways.

2010 is shaping up to be a very busy year for ISMRD. We are taking the opportunity to host an ISMRD family meeting in the USA immediately following the WORLD Lysosomal research meeting, in Las Vegas, on 19 and 20 April 2011. In addition to this, we are making plans for a scientific and family meeting in Europe around the middle of 2011. This will be an important opportunity to reach out to other Glycoprotein storage disease families beyond the mainly English speaking membership we currently have.

**Best wishes**  
**John Forman**  
President ISMRD



## ISMRD Financials in the last 12 months

*By Mark Stark*  
*Vice-President, Treasurer*  
*ISMRD*



ISMRD has gone through a significant transition in the last year; we no longer have an executive director to maintain our accounts, although we have retained our accountants to continue with required filings and to maintain our accounts. We have established new governance for our finances that gives multiple board members immediate access to our financial information.

**Financial Transactions:** In the past, expenses and deposits were done manually (checkbooks and deposit slips), and the actual status of accounts was updated periodically by a single board member. We have now set up all our accounts in Bank of America, the information from this account is provided on a monthly basis to our accountants, and three board members have access to the online banking site. Most expenses are now completed through online banking, which saves postage. The regular reports on financial information are available to all board members on our secure website.

**Account status:** Thanks to a great fundraising year by all the ISMRD families, and significantly reduced expenses, we have over \$45,000 USD in the ISMRD account. Our expenses for our accountant, financial filings, website hosting, postage and publications are about \$15,000 per year. Therefore, we have a healthy balance

which can be used to raise awareness for our children's diseases, and to contribute to research to cure these diseases. Our 2010 budget was approved in our most recent board meeting, and ISMRD will support these projects while keeping a healthy balance sheet.

**Non-Profit status:** United States law requires annual filing as a non-profit entity. ISMRD is current on all required documentation and fees. We are registered with the State of Maryland as a 501(3) (c) entity.

**Tax and audit filings:** We have filed all required United States tax documents, including required employer tax documents for our Executive Director, who was a paid employee through the early part of 2009. We also completed an audit for 2007. After consulting with tax experts in the United States, we received the advice that audits are not required for non-profits with income under \$100,000 USD. Audits in the US are quite expensive, so the board determined not to complete audits for 2008 and 2009.

**Document storage:** As Treasurer, I am retaining hard copies of all financial records, tax filings, etc. at my residence in California, United States. In addition, we are retaining electronic copies of all documents on our secure web site



## ISMRD Projects in last 12 months

*By Jenny Noble  
Secretary ISMRD*

After what has felt like the longest 12 months in ISMRD's 10year history, we are finally in a position to move forward with our mission of support, information and research. We have been working on the following projects:

**Website:** This has been a yearlong project which has seen the website go through a small makeover with more updates to happen now that we are live on the new server. Information for ML II has been added and we are currently working on the Natural History Study pages which will be included over the next few months. Our grateful thanks go to the New Zealand Organisation for Rare Disorders for the many hours of work that has taken place to ensure a smooth transition and who are our financial sponsors for the website.

**Café:** The café has been upgraded to a newer version and has also been transferred to the new web server. We hope there will be no teething problems but if you should experience a problem please contact Jenny Noble [jenny.noble@xtra.co.nz](mailto:jenny.noble@xtra.co.nz) the café web address is [www.ismrdcafe.org](http://www.ismrdcafe.org) there is also a link on the home page of the website.





**Publicity Materials:** We have been developing a set of publicity materials for displays at conferences and to help support families hosting fundraisers. I want to thank all the families who have allowed us to use their children's photos on these materials. Having publicity materials will allow us to increase our impact at conferences, and ensure that families holding fundraisers have appropriate information to share within their communities. At the present time we will be printing one banner but once funding allows we will eventually have two, to be held in different parts of the world. The brochure will be able to be downloaded from the website. However, if large numbers are needed for a fundraiser we will get them printed professionally, depending on available funding.

Our very first display using the new materials will be at the 2010 International MPS and Related Diseases symposium being held in Adelaide in June. We have a dolls house that has been donated and will part of a small fundraiser for ISMRD during the symposium. Thank you to Carolyn Paisley-Dew for organizing this.

**Fundraising:** Raising funds via charitable grants has always been difficult for ISMRD and we have had to rely heavily on families hosting fundraisers to help meet some of our upfront costs and special projects.

We hope to be able to take some of the load off our families and be more proactive with our grant writing. Currently we have 4 grants submitted with requests to help cover our day-to-day operations and publicity materials.

The next major grant submission to be made will be for the translation of our website. This project has been on our agenda for quite a few years and is being brought forward as something we need to do urgently. The cost to translate into 4 languages is approximately \$27,000US and although we are submitting a grant, we would welcome any suggestions for other possible avenues of funding.

To help meet some of our up-front costs over the last 12 months, I want to acknowledge the funding support that has come directly from NZORD who are sponsoring the website and café, LDNZ who are meeting the costs of printing and postage and Mark Stark who is covering the costs of managing our Accounts. Without this very generous support ISMRD would very quickly go through the funds we currently have in the bank.

***It has been brought to our attention that some families may not have been acknowledged in the appropriate way during the transition process. If we have missed out anyone please accept our sincere apologies. It was extremely difficult to keep a track of what everyone was doing and where the funds were coming from. If you are a family member who has not received our letter of thanks and receipt for tax purposes we would very much like to hear from you. If you do plan to have a fundraiser please let us know so that we can provide you with brochures and any other support that you might need. We are one big family working towards a common goal of research, treatments and ultimately therapies.***

Our very grateful thanks must go to each and every family who has raised funds for ISMRD whether it is a donation via the Just Give site or paypal or via the fundraisers such as the Rock for Dakotah, Lakewood Charity Dinner, Ethan's Slide, Glide and Ride bash, CDs that Pam Tobey has been selling, or the Kimmet family monthly donations. We are very grateful for the funds that are sent in. Without this wonderful support we would not be able to move our mission forward.

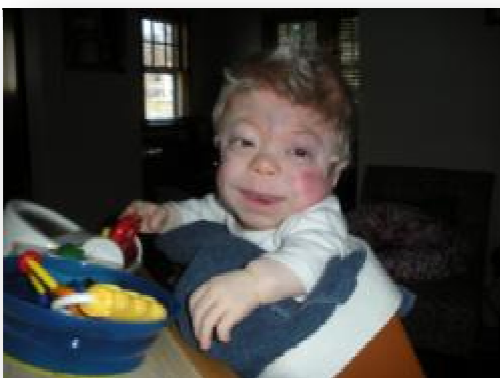


*We would like to welcome the following families  
to our penguin Colony.*

- April Riley whose daughter Madison has ML II and lives in the USA.
- Riya Jaishee whose daughter Amilie has ML II and lives in Germany.
- Michelle and Mark Vaughan whose daughter has Alpha Mannosidosis and lives in Australia.
- Lucinda Crompton whose daughter has Alpha Mannosidosis and lives in Texas.
- Lorenzo Lo Monte whose cousin has Alpha Mannosidosis and lives in USA.
- Stephania Semova whose son Evtim has ML II and lives in Bulgaria

**Ethan's Slide Glide and Ride 2010: the Journey to Our  
First Fundraiser**

*By Tara Finne  
Proud Mum of Ethan*



Our son, Ethan, is two years and two months old and has been diagnosed with ML II. He was diagnosed at one month of age after two misdiagnoses and several complications associated with birth. I was given information about ISMRD at the time of his diagnosis, along with countless other pamphlets, articles and web site addresses. All of which was terribly overwhelming to a new parent just starting to try to understand this rare disease and its ramifications. I put the papers aside and decided I just wasn't ready to tackle reading any of it for a while. After about two months of trying to keep

up with doctor appointments, home medical equipment, as well as Ethan's daily care, (which was quite difficult in the beginning) I decided I might try to read one or two of those pamphlets. I sat with the papers and went through them, one after the other, weeping as I went, still trying to grasp why my son had to suffer so much. When I finished reading, I threw them all away.

Information on ISMRD was included in that stack of paper and like the other information; I read it and threw it away. Several months later, I finally got the courage to look up an email address for another parent from the MPS web site and write a note. She was kind and gracious and led me to the ISMRD site and the penguin café. I introduced myself, my husband and our son and within hours I had a new family. So many other parents reached out to me and for the first time since Ethan's diagnosis I felt a sense of relief and peace. Here were other parents who understood us, understood our daily struggles and fears, our grief and pain, and here at last was a place for us to connect.

Ever since that first contact I have been a loyal Penguin, and visited the café often either asking for support or supporting others. This year for Ethan's second birthday, I wanted to do something to help one of the organizations that has benefitted Ethan on his journey, but I couldn't decide what to do or for whom to do it for. I then received an email from Jenny Noble talking about Rare Diseases Day on February 28, encouraging all of us to spread the word about our kids and their illnesses through fundraising or events. My husband and I decided to take up the challenge but without setting any real type of financial goal so as not to pressure ourselves. Admittedly, I was hesitant at first. I felt nervous about talking about Ethan's disease to strangers, afraid my emotions might not be able to handle it, but with support from my husband we decided to go for it. Scott, my husband works with a local park that provides winter recreation near our home in Minnesota. Together with the park we decided to promote Rare Diseases Day and honor Ethan's birthday with a special simple event. The idea was that we would invite family and friends to a day of winter recreation, including snowboarding, cross country skiing and snow-tubing, and the proceeds from all individuals specifically invited would benefit ISMRD.

Scott and I sent emails to all family and friends both local and non-local, inviting everyone to either come to the park or donate separately. On the day of the event, we had several people come to enjoy the park and donate to our cause. The bulk of the money we raised however came from individuals donating separately by sending checks directly to ISMRD, to us, or donating on line. With the power of the internet, even those not able to attend our day of winter fun were able to participate. It may have been easier to simply solicit funds on line without holding an event, but I think the fact that we did so is incredibly important. We demonstrated that we were willing to create a special day for Ethan and ISMRD, and were not simply asking for cash through a mass email. All told, we rose close to \$1500US for ISMRD, far more than we ever imagined for our first attempt, and it was far simpler than I originally anticipated. We are also incredibly excited to try again next year!

For anyone out there on the fence about fundraising, understand that even the smallest ideas can yield impressive results and every dollar counts. Our children's' diseases are rare, and it is necessary that we inform as many people as possible in order for more research to be done and treatments to be created. Fundraising can accomplish this and also be of vital assistance to ISMRD, an organization that does so much for our kids and their families. Personally, ISMRD and the Penguin Café helped me to feel a bit more alive again, as I adjusted to Ethan's diagnosis. I encourage all people whose lives have been touched by lysosomal storage diseases to get involved in any fundraising effort regardless of how large or small. Together we can all make a huge difference in the lives of our children and continue the excellent work and mission of ISMRD.

## Dr Sara Cathey and Jenny Noble were interviewed during Natural History Study in New Zealand and Australia

During Dr Cathey's visit to Australia and New Zealand in November 2009, Dr Cathey and Jenny Noble did an interview with Shine TV in New Zealand. To see the interview go to the link below:



<http://www.youtube.com/watch?v=9tHJIIONHE>



## Pam Tobey Sings and Records a CD to raise funds for ISMRD

The family of Autumn Tobey has had an ongoing fundraiser for several months now. According to Pam, her pastor asked her to make a CD as she has sung in church since she was 5 years old. Pam laughed as she is NO professional, but did agree to do this if the sales of the CDs could be donated to ISMRD. Expecting to sell just a few, Pam and some of her friends got to work. She found a cheap studio to record in and paid for those sessions and a master copy.

Then she bought blank CDs and CD cases. Friends of the family started burning copies with the ISMRD penguins lasered on the disk and Pam wrote a dedication in honor of Autumn and in memory of those who have earned their angel wings. She also wrote about ISMRD and how it has brought us all together as a family. The title is Songs of Faith and Encouragement. That was all incorporated into the front and back covers of the case. To date they have sold around 300 copies at \$10 each and are still getting a few orders. So that means around \$3000US has been raised so far. What's more, because of the CD Pam has been invited to sing at several churches and community functions and is able to tell about ISMRD's special children and the organisation.







*Early this year ISMRD was invited to become a partner in the global genes project celebrating world rare disease day. We think this is an exciting project which will work well for ISMRD in 2011 to raise awareness for our group of rare diseases. Look out for our awareness and fundraising package for the 2011 campaign.*

## HOW DID THE GLOBAL GENES PROJECT COME ABOUT?

As part of World Rare Disease Day 2009, a video began circulating on You Tube that was developed by a rare disease parent advocate. The video showed the natural connection between jeans and genes.

Using that video as inspiration, a group of individuals and rare disease organizations decided to take this connection to the next level by creating the Global Genes Project, a grassroots effort to use jeans to raise awareness for rare genetic disorders.

This group has grown and continues to add individuals and organizations that want to be involved. Our hope is that the rare disease community as a whole will view this initiative as an opportunity to build unity around this important cause. Creating a platform for collaboration, while building awareness, educating and engaging support from the general public. To learn more about this new group visit: <http://www.globalgenesproject.org>

## NIH Announces New Program to Develop Therapeutics for Rare and Neglected Diseases



**Bethesda, Md.**, Wed., May 20, 2009 — The National Institutes of Health is launching the first integrated, drug development pipeline to produce new treatments for rare and neglected diseases. The \$24 million program jumpstarts a trans-NIH initiative called the Therapeutics for Rare and Neglected Diseases program, or TRND.

The program is unusual because TRND creates a drug development pipeline within the NIH and is specifically intended to stimulate research collaborations with academic scientists working on rare illnesses. The NIH Office of Rare Diseases Research (ORDR) will oversee the program, and TRND's laboratory operations will be administered by the National Human Genome Research Institute (NHGRI), which also operates the NIH Chemical Genomics Center (NCGC), a principal collaborator in TRND. Other NIH components will also participate in the initiative. The NIH defines a rare disease is one that affects fewer than 200,000 Americans. NIH estimates that, in total, more than 6,800 rare diseases afflict more than 25 million Americans. However, effective pharmacologic treatments exist for only about 200 of these illnesses. Many neglected diseases also lack treatments. Unlike rare diseases, however, neglected diseases may be quite common in some parts of the world, especially in developing countries where people cannot afford expensive treatments. Private companies seldom pursue new therapies for these types of illnesses because of high costs and failure rates and the low likelihood of recovering investments or making a profit. **To read more visit:** <http://www.genome.gov/27531962>

## LONNIE'S (FINALLY SUCCESSFUL) JOB

*By his very proud  
parents*

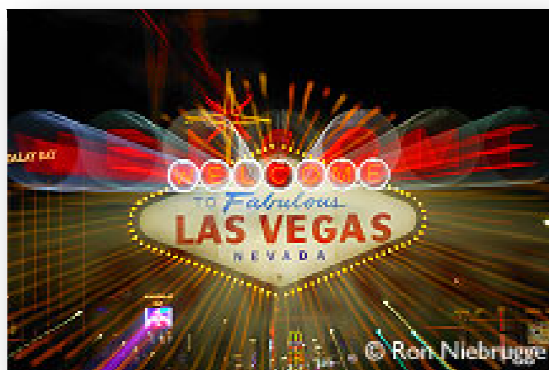


When Lonnie graduated from high school in the spring of 2007, he had no immediate job prospects. There were interviews with job counselors at LARC and the Florida Department of Vocational Rehabilitation, but jobs were scarce, and Lonnie was told, over and over again, that when jobs are hard to find, the disabled are the last to be hired (and the first to be let go, if one has a job).

So Lonnie did volunteer work at his sibling's elementary school and at his church, and during the holidays in 2008, he rang a bell for the Salvation Army.

When the summer of 2009 arrived, Lonnie and the members of his family were growing more and more frustrated. Despair and loss of hope were setting in. Then a miracle appeared from Vocational Rehabilitation in the form of a two-week culinary arts program co-sponsored by Helping Hands on Education and Hyatt Hotels. Lonnie would be away from home for two weeks for the first time ever, living in a hotel with an unknown roommate. Would he make it? Would he drop out? The answers were yes to the first question and no to the second. He made friends, learned a lot, had fun, and graduated. He never even got homesick! His family was proud of him, and he was rightly proud of his accomplishment.

Then came the visits to restaurants, and the ignored applications, until Amy Hopperstad and Don Jernigan, two of Lonnie's high school special education teachers stepped in. Even though Lonnie had been out of school for two years they admired his fortitude in the face of his difficulties so much they went to work to help him. After visiting a chef's meeting in Fort Myers, Mr. Jernigan met Chef Chris, at a country club called Heritage Palms, who agreed to give Lonnie a chance. The job has had its bad moments, such as when Lonnie was placed on unpaid probation because of not working fast enough. But now the executive chef understands that Lonnie suffers a lot of pain in his arms, legs, and back after standing and cutting vegetables and fruit for a few hours. Lonnie is being paid again, and he will be learning new tasks that will be easier for him. Hopefully, a career is born!



# **ISM RD ANNOUNCES**

## **FAMILY MEETING**

### **LAS VEGAS**

**19<sup>TH</sup> - 20<sup>TH</sup> FEBRUARY 2011**

Mark this date in your Calendar.

Our family meeting will be held directly after the WORLD Lysosomal meeting which is 16<sup>th</sup> -18<sup>th</sup> February 2011.

We are in the very early stages of planning and as we have more information we will be updating you through our website, café, face book and mail outs.

*Sadly we mourn the loss of the following children*



Olivia Armand - ML II 21-12-2004 – 11-12-2009

Faith Webb - ML II 7-1-1996 – 14 -2-2010

Gabriella Giannone - ML II 28-2-2010



**Robert Cathey – Husband to Dr Sara Cathey**

Sadly we all mourn the loss of Sara's husband Robert who passed away suddenly on 29<sup>th</sup> March 2010.

*At the time of writing this newsletter the following people are in hospital, recovering or waiting for surgery*

Jenny Klein – Jenny has fractured her hip and is in considerable pain

Hannah Voltz – respiratory infection

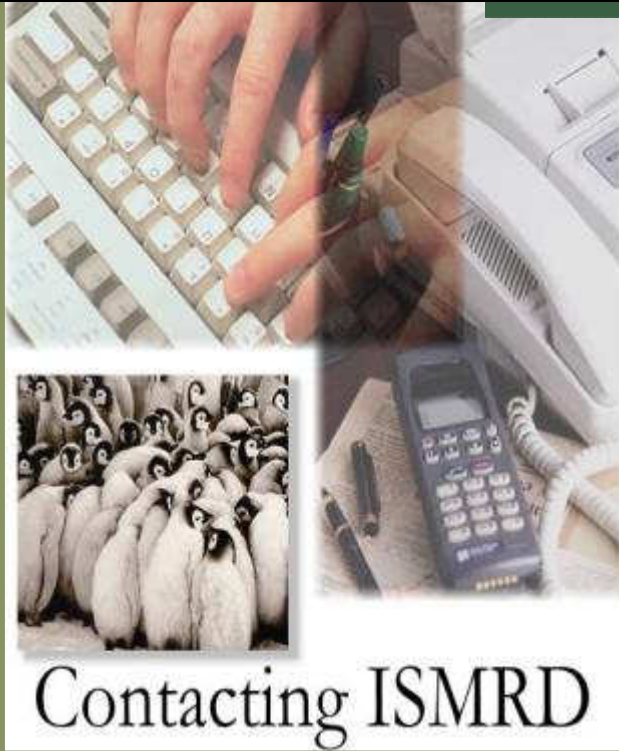
Autumn Tobey had a shoulder replacement and is now home and recovering well.

Sarah Noble – Open Heart surgery to take place within the next 3 months.



*We wish you all a speedy recovery*

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



ISMRD Board of Directors

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

**Secretary:** Jenny Noble

Directors

United States: Barbara Burton, MD | Andrea Gates |  
Pam Tobey

Australia: Carolyn Paisley-Dew

Founded in March 1999

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Footsteps In The Sand



*One night a man had a dream.  
He dreamed he was walking along the beach with the LORD.  
Across the sky flashed scenes from his life.  
For each scene, he noticed two sets of footprints in the sand:  
one belonging to him, and the other to the LORD.  
When the last scene of his life flashed before him  
he looked back, at the footprints in the sand.  
He noticed that many times along the path of his life  
there was only one set of footprints.  
He also noticed that it happened at the very lowest and saddest times of his life.  
This really bothered him and he questioned the LORD about it:  
"LORD, you said that once I decided to follow you,  
you'd walk with me all the way.  
But I have noticed that during the most troublesome times in my life  
there is only one set of footprints.  
I don't understand why when I needed you most you would leave me."  
The LORD replied:  
"My son, My precious child, I love you and I would never leave you,  
During your times of trial and suffering,  
when you see only one set of footprints, it was then that I carried you."  
**Author unknown***



# 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
  - Any other ideas, or 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 & contact details**

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

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

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

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

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

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

