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

- **December 30th 2011 – advising Pam Tobey you want assistance with Mercy flights.**
- **March 31st 2012 – Deadline for booking your accommodation with the Crowne Plaza, and registering with ISMRD.**

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 President's Desk

By John Forman
President ISMRD

Hello to the ISMRD network.

Much of the ISMRD board's time during this year has been devoted to planning for the July 2012 conference in Charleston, South Carolina. We are pleased with the way the program is shaping up and we feel confident families will get great benefit from attending this event. With the natural history study clinics taking place on the Friday before and the Monday after our weekend family meeting, and with a scientific meeting being held from Thursday evening to midday Saturday, there is certainly going to be plenty happening there.

This newsletter contains more detail of the conference events and registration information, and you should also be receiving other notices of this mailed out to you as a reminder. We look forward to seeing many of you there. It will be great to meet you again, or for the first time for new families. Please remember to register early to ensure accommodation during this busy time, and to get your request for flight assistance in on time.

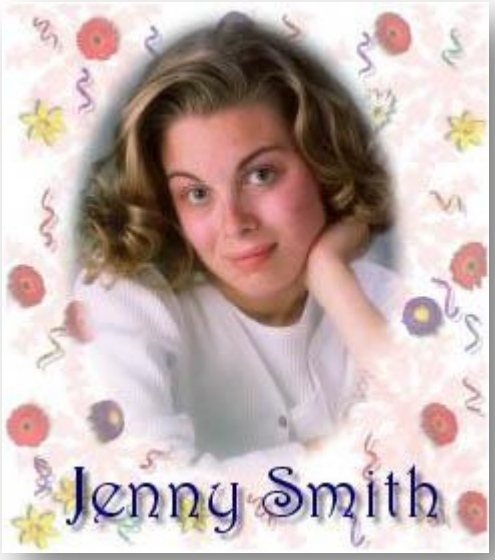
There is more good news in this issue with ISMRD winning one of the Patient Advocacy Leadership awards promoted by Genzyme. The grant received will go towards the publication of clinical care guidelines for Mucolipidosis Type 2 and 3. There are many complications with both these disorders. This project will ensure the knowledge and experience of families plus the expert analysis by Dr Sara Cathey and her team will become available in the published medical literature to assist doctors and families worldwide in the care of the complex symptoms of these diseases.

Of course research into our 9 diseases is a major part of ISMRD's mission, and we are exploring ways in which we can help more research to occur. Providing funds to develop clinical care guidelines is one very practical way we can have an impact on disease knowledge and improve clinical care. We know many families would like to support more research and we are having discussions with our professional advisory board to explore ways in which ISMRD can have an impact on research despite our small numbers and limited funds available. We plan to outline some of these ideas to you at the Charleston meeting.

And just to show that good news can come in 3s, Scandinavian biotech company Zymenex has recently announced the successful completion of their phase 2a clinical trial of an enzyme replacement therapy for Alpha-Mannosidosis, and the commencement of the 2b phase of their trials. We look forward to more news and we are working on having a presentation on this trial at the Charleston meeting.

My very best wishes to you all for the coming holiday season. From New Zealand, home of the champion All Blacks, winners of the Rugby World Cup 2011.

John Forman
President ISMRD



In Loving Memory

ISMRD received the sad news of the passing of Jenny Smith on 24th July 2011 after a serious illness. Jenny had Alpha Mannosidosis. Jenny's journey through Bone Marrow Transplant is on the ISMRD website.

Jenny's mum Dianne has written a little bit about what happened to her.

From July to September, 2010 both Jen's kidneys failed. We took her to Mayo Clinic Hospital in Phoenix, Arizona for a kidney transplant. Jen had dialysis 3 times a week, 4 hours per session, while the preliminary tests were done at Mayo. Jen's dad, Steve, wanted to be the first person tested to see if he was a match -- and HE WAS! The transplant was scheduled for January 5, 2011.

However, after the transplant, Jen never flourished. She just continued to get weaker, sicker and never had an appetite. This continued until June 9 when an MRI showed a cancerous tumor that encased 75% of her abdominal aorta. It was a sarcoma (which is a terminal tumor) and it was inoperable. Jen's condition declined until her death on July 24.

We would be so happy if everyone at ISMRD remembered Jen for the brave, kind, loving, and wonderful person that she was. Her courage and dignity were second to none.

Was it you?

A Butterfly flew by my window today
Was it you?
A cool Breeze kissed my face today
Was it you?
The sun warmed my soul today?
Was it you?
I see you every time I close my eyes
You are in my every thought
You are a quite part of me
I Love you

The Will to Live – By Paul Murphy

Courage, determination and the will to live. These are the attributes I think of when recalling Jenny Smith's struggle to overcome Alpha Mannosidosis. Indeed, these attributes characterized Jenny's family: Dianne, her mother; Steve, her dad and Jason, her brother. So, it was with shock and sadness that I learned recently that she had passed away this past summer; not because of Mannosidosis, but from a cancerous tumor in her abdomen that proved inoperable. She was 35.

I became aware of Jenny through Dianne who found me in late 1998 via my website, "Rare Genetic Diseases in Children." Jenny had just been diagnosed, at age 22, with Alpha Mannosidosis and Dianne began the journey all of us, in similar circumstances, take: looking for support for a strange disease and desperately hoping to find a cure. Dianne's contact with me coincided with our initial efforts to form a nonprofit organization in Baltimore, Maryland whose objective was to raise awareness for Mannosidosis and similar diseases. When ISMRD formally came into existence in the spring of 1999, Jenny was already in full pursuit of a donor for a bone marrow transplant.

In June 1999, having successfully found a donor match, Jenny underwent a BMT at the University of Minnesota. Concurrently, ISMRD, now operating a fledgling web site, created a special section, "Jenny's Journal," that Dianne used to post a diary of Jenny's progress. For the next two years Jenny's recovery became very traumatic for her family, as complications from Graft vs. Host Disease (GVHD) negatively impacted her health and every encouraging development seemed offset by two offsetting setbacks. Her struggle was such that her family feared she may not survive and, in fact, on a couple of occasions she came close to passing away.

In late spring of 2000 we were privileged to meet Jenny and her family in Minnesota, where they resided. By "we" I mean myself, my daughter Taryn, her mother, Debora and John Forman. All of us had travelled to Minneapolis to attend the "6th International Symposium on MPS & Related Disorders." John, Debora and I represented ISMRD's "related disorders," as members of our organization's Board of Directors. It was an extremely positive conference, which enabled us to do some very critical networking on ISMRD's behalf.

Knowing we would be in Minneapolis, and in the vicinity of the Smith's home, I prearranged a visit to see them and meet Jenny. For all of us, it was the first time to meet another person affected by Alpha Mannosidosis (other than our own children). Likewise, the Smiths had never met another family so affected. We had a fabulous time and were able to finally place names with faces. Our visit was not without some concern, however. Jenny was very impacted by GVHD; the major visible impact was her swollen appearance. It was difficult, I admit, to reconcile the young lady I now saw with the photos of her taken pre-transplant. I could not believe that they were the same person.

Nevertheless, Jenny maintained a positive attitude throughout our stay and onward during her long recovery. She not only persevered, but seemed to thrive as time passed. This was confirmed four years later when we again saw the Smiths at ISMRD's "1st International Conference on Glycoprotein & Related Storage Diseases" in Rockville, Maryland. Jenny looked great and had lost the swollen appearance I noted in 2000. She greeted all of us warmly, especially Taryn, her fellow "aman girl!" Jenny spoke of her dreams of writing a book one day and travelling. She was full of hope and possessed of a very positive attitude, which I found remarkable for someone whose last few years were filled with such agonizing challenges. But that was Jenny: a persistent, stubborn optimism and determination to live and "beat the odds."

It is with a mixture of sadness and delight that I recall Jenny and am grateful that I was blessed to know her and her family. Many of us who live with these strange and dire diseases have had such an experience: being touched by someone and "losing" them one day. Though Jenny was not a direct victim of the disease she was born with, her life was indelibly impacted by it. We can do justice to her memory, and to others who have similarly seen their lives shortened, by supporting organizations like ISMRD that strive to find therapies and cures. Jenny would insist that we do so.

Postscript:

I have been out of touch and "out of the loop" for quite a long time; long enough that many of you who read this may not even know who I am. For those that do remember me, let me bring you up to date. Taryn, my daughter, is now 27 (yikes!) and participates in a day program in the Baltimore area designed for adults with disabilities. She, like others with Mannosidosis, has experienced a decline in her physical and intellectual abilities which provides us with concern and anxiety. Along with these are concessions we've had to make to each new "milestone:" rolling walkers, wheelchairs, adult diapers, etc. Nevertheless, she has not lost her sense of joy and her smile, which makes it all worthwhile to me. She has experienced another grave challenge, however. In the fall of 2006 she was diagnosed with Chronic Myelogenous Leukemia, which set our world on end but for which, fortunately, there is a treatment. Because of a miracle drug called Gleevec, which she must take daily, she is in remission and has no visible effects from the disease (knock on wood). Indeed, if only it were so easy with Mannosidosis; then all would be super! Take care and have a great holiday season to come. If you want to say hi, I am on Facebook, Twitter and can be reached at paul3210@comcast.net Until next time...

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 26th – July 30th 2012

The conference dates are broken down as follows:

- Scientific conference July 26th -July 28th
- 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.

Mercy Flights are again going to help us get patients to Charleston for the Natural History Study. Please e-mail Pam Toby dtobey@centurytel.net with your request for assistance no later than ***30th December 2011***

- ***Request your Mercy Flights with Pam Tobey before 30th December 2011***
- ***Book your accommodation with the Crowne Plaza before 31st March 2012,***
- ***Send your registration forms and fees to ISMRD before 31st March 2012,***

We look forward to warmly welcoming you all to Charleston South Carolina!

Natural History Study

The extension of the Natural History study will take place at Dr Sara Cathey's Charleston Office.

Your family knows a lot about one of these conditions. Take this opportunity to share your story.

Information about growth, development, physical findings, learning, medical complications, and survival will be collected from affected individuals. A clinical geneticist will collect/collate the history and perform a physical examination. A psychologist will assess learning and development. Biological specimens (blood, urine, tissue biopsies) will be obtained for biochemical and molecular analyses. The specimens are needed by researchers and scientists studying the glycoproteinoses. The compiled clinical data will become a valuable resource for families, medical care providers, and researchers. Families will be given the results of their gene mutation analyses. This is a tremendous opportunity not to be missed.

You have waited and wondered when the world would pay attention to these diseases.

We are ready! We need your help!

Conference Venue

Crown Plaza Charleston SC |Airport Convention Centre Hotel

We have secured our accommodation and meeting rooms at the beautiful Crown Plaza Hotel. The Accommodation rate per night is $109+ taxes. This rate is also available for dates either side of the conference to cater for those who need to arrive earlier due to long distance flights.

The hotel is set up now to receive your accommodation bookings. When booking your accommodation please use the following group code ***GFV.*** We encourage you to book early as July 2012 is an extremely busy time in South Carolina. ***Late bookings may not be able to be accommodated after 31st March 2012.***

Contact details for the hotel are:

Crowne Plaza Charleston Airport Convention Center
4831 Tanger Outlet Boulevard
North Charleston, SC 29418

Phone 877-503-5762

To read more about the hotel please visit. http://www.crowneplazacharlestonhotel.com/index.php

ISMRD wins Patient Advocate Leaders

ISMRD is very proud to announce that it has won a Patient Advocate Leaders Award for Innovation from Genzyme.

The purpose of this grant is to allow us to develop and publish Management Guidelines for Mucolipidosis II and Mucolipidosis III (ML II and ML III). This will be a first-ever medical paper that sets forth disease management guidelines for the complex symptoms of ML II and ML III.

Out of 55 applicants, 11 patient organizations were selected by an External Review Committee to win a PAL Award. Proposals were received from patient organizations in 25 countries. The successful applicants came from Brazil, Bulgaria, Canada, Colombia, India, Italy, the Netherlands and the United States. To read more about this grant please visit. http://www.genzymeadvocacyawards.com/

This is a very exciting first step forward and is a specific example of the new focus that ISMRD will have in coming years. There will be a need for everyone to be involved in raising funds which will be targeted to specific research, clinical care improvements and Natural History study extensions. We will be announcing more about this later and when we do we will be seeking support from families for specific funding. This is truly an exciting step forward and we hope that together we will forge a new pathway forward for these very rare diseases.

We warmly welcome to our Penguin Family.

- Theo Prasidis and his cousin Kyriakos who has Galactosialidosis and lives in Greece.
- Antonn Peach who has Sialidosis type 1 lives in New Zealand and also has two other affected siblings.
- Sarah Ali whose son Yusuf has Alpha Mannosidosis and lives in the UK
- Trish Adkins whose son had ML III and lives in the USA
- Syeda Adia Zaidi whose son has Alpha Mannosidosis

Fundraising for ISMRD – We need your help!

By Andrea Gates

Dear I.S.M.R.D/Penguin Families!

I just wanted to touch base with some of you and let you know about some of our exciting things happening with I.S.M.R.D. As you know ISMRD supports 9 diseases: Alpha Mannosidosis, Mucolipidosis II and III, Galactosialidosis, Aspartylglucosaminuria, Fucosidosis, Beta Mannosidosis, Schindler Disease and Sialidosis. Wow can you say that three times fast. I can't even say it once! I have a hard enough time saying and spelling Mucolipidosis for doctors and nurses, lol.

Anyway…I am excited that I.S.M.R.D. is 100% focused on becoming a major source of support for both families and research for treatments/cures. Through our persistence, I.S.M.R.D. is now bringing our diseases to public awareness and getting noticed by the scientific/medical community!!!

Now the exciting news…The Family Meeting/ Natural History Study will be in Charleston, SC with Dr. Sara Cathey in July 2012. This natural history study will include all 9 diseases that I.S.M.R.D. supports. This is so exciting! We will once again be able to come together in one place. For some of us it will be the first time we meet and the first time our children are face to face with kids just like them. For others it will be like a family reunion. I can still remember the first time Spencer met Matthew, Joey, Sergio, Andre, Anna and Zachie and many others. He said that he never knew there were others just like him and that he did not feel alone anymore. I couldn't have agreed more and am always thankful that I.S.M.R.D. brought us all together. That in my opinion is and will always be priceless.

As well as applying for grants, we are appealing to **families** to come up with some creative ways to raise funds, big or little. Anything you can do for I.S.M.R.D. will be wonderful. Wouldn't it be wonderful to help raise money to fund these projects for our "kids"? Please don't worry about getting started or think you can't do it. You can! We are all parents with similar challenges and busy lives.

We need to all pitch in and help I.S.M.R.D. to reach our common goal of having the best life for our kids and hopefully a cure. As for my family, we will be hosting a Poker Night in the next few months. It has been a couple of years since our Charity Dinner and it's time for us to again step up to the plate and help the organization that has opened up doors for our family and introduced us to so many of you. We need to again "pay it forward". So if you think ISMRD has helped you and your family in any way then it's time to "pay it forward' and give back.

Please think about raising funds for I.S.M.R.D. I can only speak for myself but I don't know where I'd be without the families I've met, the doctors and researchers I've been in contact with, the support of parents that know exactly how I feel and all of my questions that have gone unanswered before I.S.M.R.D. We, me, us, the board, are all here to help families that may need it. We are in this battle together. You can reach me by email, face book or directly on my cell phone 562-884-2366.

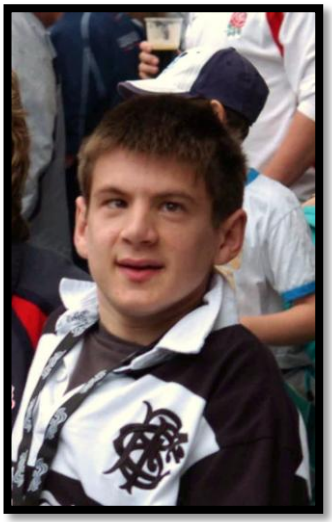
Love to All My Penguin Families!

Chris Leonard gains some independence.

By Jean Leonard

Chris's mum Jean has written an update about Chris who has Fucosidosis and lives in the UK. John and Judith Forman met Chris and his family along with Martin and Sonja Woolley while they were on holiday earlier this year.

Chris is nearly 20 and has completed 1 year in the supported learning unit at his local college and that has gone very well. He is learning catering, and IT and living skills. Chris moves around using a rollator frame, he can also sit on this if tired and store stuff in the seat. A great deal of his friends from school have gone away to residential college, however Chris has made new ones here in Twickenham.



We have a good self-directed support budget from the local authority and the power to use this for carers/respite etc. This long summer holiday he has been out and about with me or other student carers to support him. He is having a great time. He also has a small job (with another support carer) recycling bouquets of dead flowers, he really works hard at this. He also does the shredding for a local office. He misses the rugby during the summer.

Chris still loves going to cafes, bowling, youth club & seeing friends. We (mum & dad) are just tolerated.

Zymenex enzyme gets EU Orphan Drug Designation Approval

October 3, 2011 The European Commission (EU) has on September 27, 2011 granted ACE BioSciences A/S (a subsidiary of Zymenex Holding A/S) Orphan Drug Designation for its recombinant human enzyme Galaczym, for the treatment of the rare, lysosomal disease Globoid Cell Leukodystrophy (Krabbe Disease). Orphan designation qualifies the sponsor of the product for fee reductions, protocol assistance, centralized application procedure and access to 10 years marketing exclusivity once authorized, according to the Orphan Drug Regulation. Zymenex has previously received Orphan Drug Designation in both the EU and US for two other lysosomal enzyme products Metazym for the treatment of Metachromatic Leukodystrophy and Lamazym for alpha-Mannosodisis, which is presently in Phase 2 clinical trials. Read more go to www.zymenex.com/composite-80.htm

Positive Clinical results on Phase 2 Clinical Trial for Alpha Mannosidosis

October 17, 2011 The biotechnologically derived human enzyme rhLAMAN (LamazymTM), which is produced by the Danish biotech company Zymenex and developed for the treatment of patients suffering from the rare lysosomal storage disease alpha-Mannosidosis, has successfully completed a Phase 2a clinical trial.

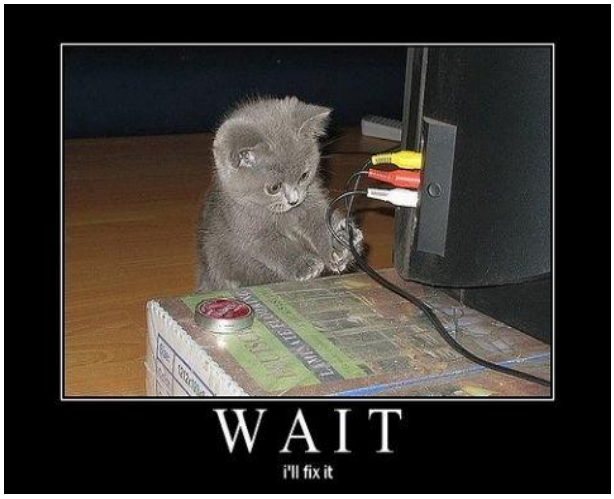
"9 patients aged 7 to 18 years were recruited to the Phase 2a clinical trial from European hospitals and each week flown to Copenhagen, Denmark to be treated here at the Department of Clinical Genetics, Copenhagen University Hospital", says Chief Physician Dr. Allan Meldgaard Lund MD, who is the Principal Investigator and treating physician. "There is an unmet need for the patients suffering from this disease and we are very encouraged by the results we see."

The patients were divided into two cohorts receiving doses of 1 mg/kg or 2mg/kg respectively each week. Apart from finding the minimum effective dose, the goal of the Phase 2a trial was to demonstrate that the enzyme was effective and improved the patient's condition. There was a clinically relevant improvement in the parameters measured and thus the goal has been achieved.

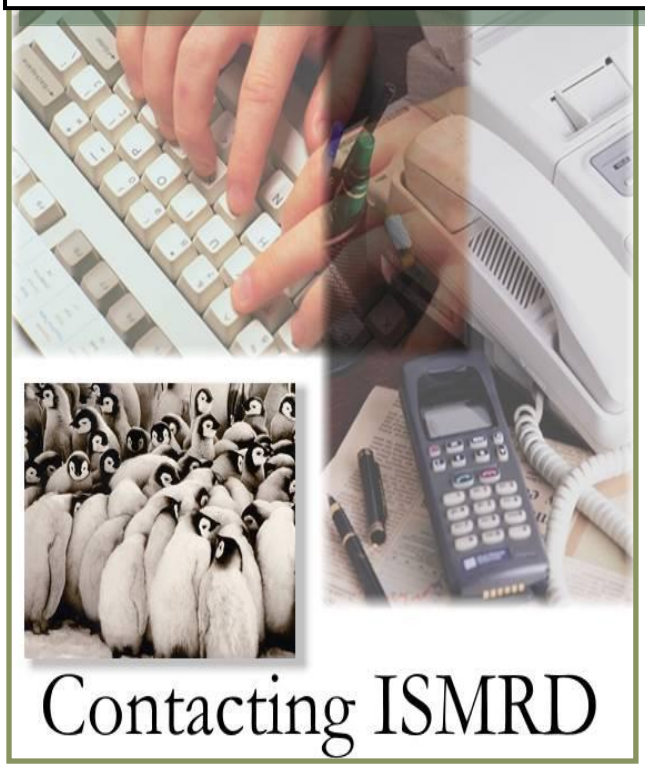
This now allows the 9 patients to be moved forward into a 6 -month Phase 2b clinical trial, where the aim is to verify the minimum effective chosen dose of 1mg/kg, by monitoring the effect on chosen composite efficacy endpoints in order to confirm the clinical effect of repeated weekly i.v dosing.

"The positive results of the clinical trials are due to the collaborations over the years between the Zymenex team and many leading scientific and clinical colleagues from different European countries in the joint EURAMAN, HUE-MAN and presently ALPHA-MAN EU Framework projects," says Dr. Jens Fogh, President and CEO of Zymenex A/S. "We have spent 10 years developing this product and are now near to our goal, which is to provide an enzyme replacement therapy available for ALL alpha-Mannosidosis patients."

To read more about this exciting development for Alpha-Mannosidosis visit www.zymenex.com/composite-81.htm



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

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Thank you for your support!

ISMRD wishes to acknowledge all these wonderful people who have donated funds to ISMRD during 2011. Without this very generous support we would struggle to meet our day to day operational expenses.

Kimmet Family, Vincent O'Connell, Pam and David Tobey, Philip and Carolyn Holzman, Peggy Wilson, Carolyn Paisley-Dew for the donation of a dolls house, Mark Stark, Woolley family, Global impact-applied materials, Blair and Carolyn Anderson, Leigh Valley ch of music teachers, Bret and Jackie James, The London Tea room, Lee Anne Ellison, Richard White, United way of central new Mexico, Marion Abdullah, Allan and Su Lane, highmark-blue shield, Allan Thomson,

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: _____

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