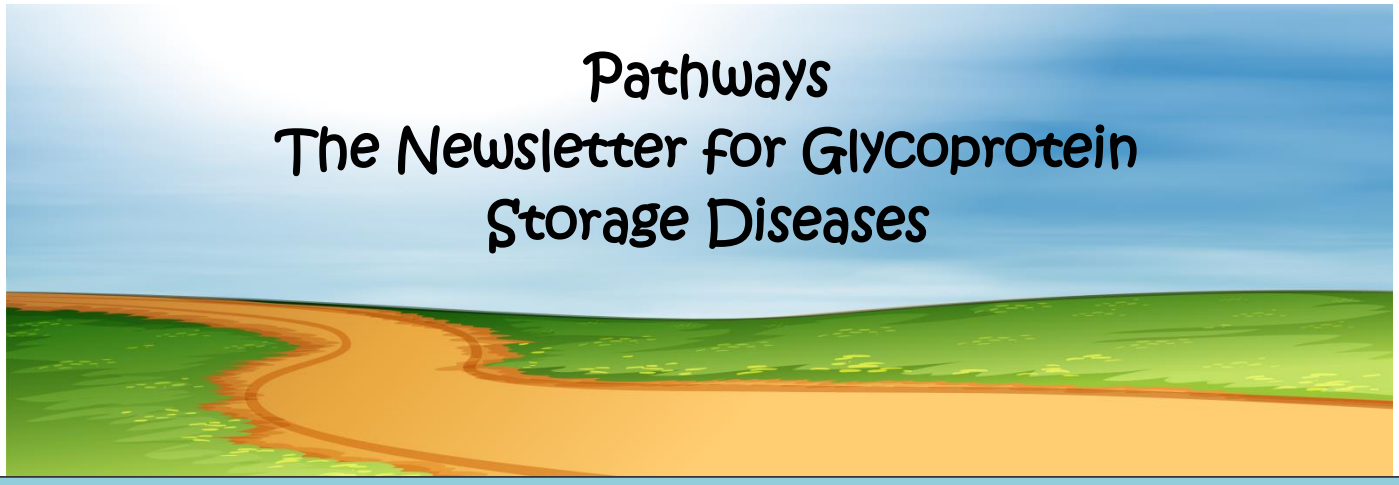

ISMRD, a 501 © not-for-profit organization, FEIN 53-2164838 | website www.ismrd.org

Our Mission and Vision

ISMRD is the leading advocate for families world-wide affected by Glycoprotein Storage Diseases.

Through partnerships built with medicine, science and industry we seek to detect and cure these diseases, and to provide a global network of support and information.

We seek a future in which children with Glycoprotein Storage Disease can be detected early, treated effectively and go on to live long, healthy and productive lives.

ISMRD supports the following disorders

Alpha Mannosidosis, Aspartylglucosaminuria, Beta Mannosidosis, Fucosidosis, Galactosialidosis, Mucolipidosis II alpha/beta (I-Cell Disease), Mucolipidosis III alpha/beta (Pseudo-Hurler Polydystrophy), Mucolipidosis III Gamma, Schindler Disease and Sialidosis

https://www.facebook.com/groups/82945687520/

https://twitter.com/ISMRD

Donations

ISMRD is a 501(c) charitable organisation based in the United States serving a global constituency. We provide our services, which include our newsletter, website, outreach activities and support of research, without requesting monthly dues or any other financial restrictions. We gratefully accept donations that will enable us to continue toward our goal of a future free of the tragic consequences of Glycoprotein Storage Diseases.

Donations can be made via our website using

PayPal Secure Payments

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Contact Us

The International Advocate for Glycoprotein Storage Diseases

20880 Canyon View Drive
Saratoga
CA 95070
USA

email: info@ismrd.org

From the President's Desk

By Jackie James
President, ISMRD

We have an exciting and very busy newsletter for you this March! I would like to start off our good news by introducing and welcoming our newest board member, Paul Wagner. Paul is the proud grandfather of Ellie Kenny, who has ML II. Many of you that attended the conference in Saint Louis may remember sweet Ellie and her parents. As well as agreeing to join us as a board member, Paul has also agreed to undertake leadership of our fundraising committee. With Paul's past experience with marketing and business, I believe he will be a tremendous asset to ISMRD.

Our upcoming conference in Rome is taking shape and we are getting very excited to host our very first meeting in Europe! Jenny Noble is working tirelessly to get all the arrangements worked out and it is shaping up to be a wonderful, informative time for all those that attend. We are very excited to branch out and reach physicians, researchers and families in Europe that may never have been able to attend previous meetings in the states. We hope that many of you can join us in Rome.

ISMRD is also very excited to announce that our ML research initiative has awarded Dr Sandra Pohl and her team a research grant looking into treatment of Osteoporosis in Mucolipidosis. Personally, I am very excited to see the results of this study and hope that Dr Pohl will be able to find relief for our kids that suffer so much with bone pain.

More exciting news is that ISMRD families have been invited to participate in a study conducted by Oregon State University. The University is conducting a study on the quality of life among adults with rare diseases. We have all of the information, along with a link to the study listed in this newsletter. Please take part.

I would like to take some time to introduce the Huber family to our ISMRD family. Evelyn Huber has written an introduction to her and her sweet daughter Franziska, who has ML II. We are so happy to welcome you to our family and hope to meet you soon – maybe in Rome?

I would also like to mention my good friend Denise Crompton and her amazing and inspiring book Diagnosis: Rare Disease. Denise has penned a couple of paragraphs in our newsletter about her book. I am very proud of Denise and all she has accomplished in helping families cope with the ups and downs of having a child with a rare disease. I'm also proud to be one of the families that Denise speaks about in her book. I recommend this book to anyone that is dealing with a rare disease.

Rare disease day 2017 has already come and gone! Families and organizations all over the world recognized the day in many different ways. Our new fundraising committee will be working hard on how we can recognize this special day next year.

We have listed an article from Medicini Clinica on how to progress from pediatric health care to adult health care. This is a transition that my daughter Anna has just experienced and it is definitely something that is easier said than done! There is a fee to view this article.

We would like to ask families to notify us if your child is facing illness or surgery, as we would like to send them a greeting in our newsletter. Without your permission we are unable to do so. We would also like to list condolences for anyone that loses a loved one. Any articles on your family that you would like to send us to publish in the newsletter are always appreciated.

Thank you to each of you that donated to ISMRD this past quarter. Your generosity is so very much appreciated. On that note, I would like to acknowledge my aunt, Joyce Blesi Knaus. In early January of this year, Aunt Joyce passed from this world. Her family knew how much she loved my daughter and her great niece Anna, and how she longed to see a cure for Mucolipidosis. In lieu of flowers, her family requested that people donate to ISMRD. I would like to thank all the family and friends of Joyce that donated and for the generosity and thoughtfulness of her daughter Chris Knaus Black who made it all possible.

I wish each of you a wonderful Easter, and I hope to be able to see many of you at the conference in Rome later this year.

Jackie James
ISMRD Board President
Mom to Anna, ML III

Denise Crompton signs copies of her book, Diagnosis: Rare Disease,
at the 2015 ISMRD Conference in St Louis, Missouri

Register now!

Planning for our conference is slowly taking shape. We hope that in the next few weeks we will be able to provide some of the program content. The Scientific committee is working hard on this.

Last week we opened the Conference Registration and Accommodation. Within 24 hours of Registration being open we received our very first registration. Way to go Juanita and Jess-Rose Van Dam in Queensland, Australia!

Our Fundraising committee is working on a fundraiser for families who want to sell items and raise funds towards their own accommodation. We will provide you with the details soon.

Registration: **Go to** http://www.ismrd.org/fifth-international-conference/registration

All fees are in USD and are per person

	Early bird registration (before 30th August 2017)	Late registration (after 30th August 2017)
Parents of affected children	**$175**	**$200**
Affected children	**$0**	**$0**
Healthy child	**$90**	**$100**
Students/junior doctors, other adults (relatives, friends, not for profits)	**$275**	**$375**
Professionals (scientists, physicians, bio-tech companies pharma, healthcare)	**$375**	**$475**

Accommodation: Go to http://www.ismrd.org/fifth-international-conference/accommodation for details. You are now able to register with the conference hotel. Please use the link http://www.hotel-aroma.com and the Code 5GIC to book your accommodation.

Hotel room costs

- 1 person – **€135.00 per night**
- 2 people – **€155.00 per night**
- Children over 12 years – **€25.00 per night**
- Children under 12 year – no extra charge

Families needing accessible rooms. During the booking process you will come to a place where you can leave a message for the hotel. This is where you will advise the hotel of your needs. Please keep in mind there are only 13 accessible rooms so it will be first in first served.

If you have any questions please let us know either through info@ismrd.org or jenny.noble@xtra.co.nz

We look forward to welcoming everyone to Roma!!

ISMRD's Mucolipidosis Research Initiative

The Wagner Foundation

Another research grant approved as part of ISMRD's Mucolipidosis Research Initiative:

Osteoporosis in Mucolipidosis II - A Potential Corrective Approach

ISMRD is delighted to announce on behalf of all our funding partners in the ML Research initiative, the approval of another research grant.

This grant will research Osteoporosis in Mucolipidosis II - A Potential Corrective approach and is awarded to Dr Sandra Pohl who comes from the Biochemistry and Cell Biology section of the University Medical centre in Hamburg-Eppendorf, Germany.

Her work will look at how osteoclastogenic cytokine interleukin (IL-6) may represent a potential treatment of osteoporosis in ML II and ML III. It is hypothesized that IL-6 overexpression in ML mouse models is responsible for their osteoporotic phenotype, therefore making IL-6 a novel target for the skeletal system but possibly also for other affected organs.

ISMRD is thrilled with the tremendous support from so many families and groups towards this outcome, and delighted to be supporting this significant new development in research into this serious disease.

Thank you so much for your contribution to making this research possible

ISMRD is asking all our families to participate in this exciting study. If you have any questions, please let us know at info@ismrd.org

Kathleen Bogart is a psychologist at Oregon State University. She both has and studies rare disease. She is partnering with the US National Organization for Rare Disorders (NORD) to conduct the largest survey of quality of life among adults with a variety of rare diseases. The outcome of this project will be to provide information to rare disease organizations and healthcare professionals about how best to support quality of life needs.

AWaRDS Study: Adults with Rare Disorders Support

What is the study about?
In partnership with NORD, this will be the first large-scale study about the information and psychosocial support needs of people living with rare disorders. The purpose of this research study is to assess these needs, from the perspectives of people with a variety of rare disorders, to find similarities and differences across disorders. To ensure that results reflect the diversity of the rare disease community, it is crucial that as many people living with a rare disease as possible take part.

What would I do as a study participant?
There are two ways to participate. 1) You can follow this link http://bit.ly/2hWZLr2 to take a 40-minute online survey about your experiences with and information and support needs related to your rare disorder (paper forms are available by request). If it is physically difficult to respond, someone may enter your responses for you. 2) During the survey, you can opt to sign up for a second study, which involves an online focus group about the information and psychosocial support needs with others with rare disorders. You must participate in the survey in order to be eligible for the focus group, but the focus group study is not required to participate in the survey. You will be paid $20 for participating in the focus group.

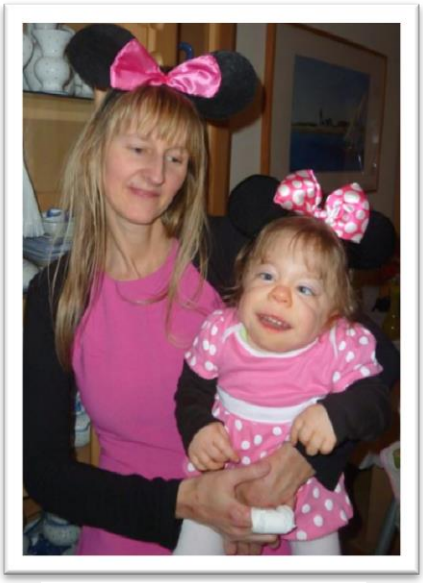
Who is eligible to participate?
You must be an adult or the age of majority in your state, be able to communicate in English, and have a rare disease or disorder or undiagnosed rare condition. Caregivers who do not have a rare disorder themselves are NOT eligible to participate at this time. A disease is generally considered rare if it affects fewer than 200,000 affected individuals in the United States or fewer than 1 in 2,000 in Europe. A list of rare diseases can be found here: https://rarediseases.infonih.gov/diseases/browse-by-first-letter. Because rare disorders are discovered and prevalence estimates change frequently, you may participate even if your disorder does not appear on the list.

What will we do with study findings?
We will send a summary of results to all participants. To help NORD, rare disorder organizations, and healthcare professions meet the needs of people with rare disorders, results will be shared through reports, conference presentations, scientific publications.

Who are the researchers?
Kathleen Bogart, PhD, Principal Investigator, Assistant Professor of Psychology at Oregon State University, studies psychosocial needs of people with rare disorders and has a rare disorder herself. She also serves on the Board of Directors of a NORD member organization. Contact her at Kathleenbogart@oregonstate.edu or 541-737-1357.

Veronica Irvin, PhD, MPH, Co-Investigator, is Assistant Professor of Public Health at OSU. She has experience analyzing information offered by support organizations.

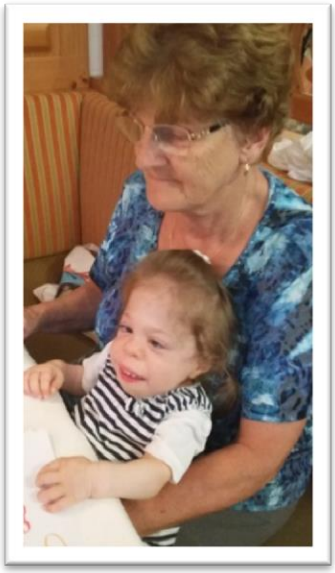
Hello from the Hubers in Austria



Franziska and her mother Evelyn

First, let us introduce ourselves. We are the Hubers. I am Evelyn. My daughter is Franziska and my mother (granny) is called Herta. We live in Austria.

On 5 June 2013, we received the devastating diagnosis of Mucolipidosis II for our daughter Franziska at the University Children's Hospital, Graz in Austria. It was a shock, but after about three weeks we began to look for help around the world. A very difficult decision was for us whether we should make a stem cell transplant or not. We decided against it; today we are of the opinion that it was and is the right decision.



Franziskz and her granny Herta

Our greatest wish is that there is a therapy for ML II, which can help our lovely girl.

Franziska is six years old; she is 80 cm tall and has a weight of 8 kg. She is a sunny girl. She loves music and likes to sing.

Our sunshine regularly gets Physiotherapy, Ergotherapy and Logotherapy. My mother and I take part with Franziska at the annual "week of therapy", organized by the Austrian MPS Society. This helps our sunshine very much. The contact and the exchange of experiences with the other families are also very important for us.

Franziska can sit unaided since 31 October 2015. Since then she has learned a lot. We are very happy that she can stand on her own with just a little help. Franziska enjoys this very much. She exercises nearly every day for her dream to make steps without help. My mum and I will do everything we can to make Franziska's dream come true.

We would like to thank Franziska's physicians, therapists and of course Mrs Weigl and her team from the Austrian MPS Society.

Evelyn Huber

HELP SPREAD THE WORD

Author Denise Crompton with her husband Bob and daughter Kelley

I decided to write DIAGNOSIS: RARE DISEASE in order to help bring about awareness about what it is like for a family living with a rare disease. I also planned to raise money for research through my royalties.

After I gathered all of the information from the families that participated, and put it all together, I realized that there is a wealth of information that can help other rare disease families in coping with the various situations that can occur in their lives while dealing with the medical community, finding support, interacting with educational systems, helping the affected find some relief from persistent pain and inevitable surgery, facing an uncertain future and the pain of saying goodbye to a loved one.

The chapters are arranged in such a way to make it easier for the reader to go to a particular topic to learn what problems others have encountered and how they have handled them. We can all learn so much from others, and I am grateful to the mothers who were willing to share their experiences with us.

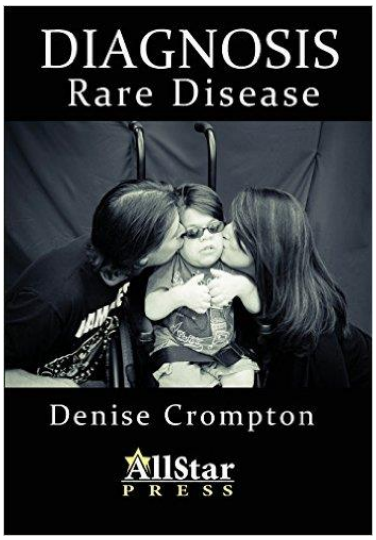
You can help spread the message! If you already have a copy of the book, you might want to get one to donate a copy to a medical center or even your own public library, as many of us have already done.

These are the links for both paperback and Ebook versions:

https://www.amazon.com/Diagnosis-Rare-Disease-Denise-Crompton/dp/1937376176

http://www.barnesandnoble.com/w/diagnosis-denise-crompton/1120351590?ean=9781937376178

Denise Crompton, Author



What's happening around the world?

RARE DISEASE DAY 2017: RESEARCH

28 February 2017 was the 10th Rare Disease Day, and saw thousands of people from all over the world come together to advocate for more research on rare diseases. Over the last few decades, funds dedicated to rare disease research have increased, but more is needed.

Research can lead to the identification of previously unknown diseases and can increase understanding of diseases. It can enable doctors to give a correct diagnosis and provides information to patients about the disease. It can lead to the development of new innovative treatments and in some cases a cure.

Rare disease research reduces costs for healthcare systems. As a result of research, a rare disease patient with diagnosed properly treated no longer needs irrelevant tests, ineffective treatment or hospital visits. In addition, research on specific rare diseases often shines a light on more prevalent diseases. Pioneering approaches in rare disease research often benefit the much wider public affected by more common diseases.

Rare Disease Day 2017 was also an opportunity to recognise the crucial role the patients play in research. Patients advocate for research, fund research, partner in research projects and participate as subjects in clinical trials.

ISMRD continues to advocate for research for our nine Glycoprotein Storage Disorders, and to raise funds for research such as our Mucolipidosis Research Initiative.

Recommendations on the transition from paediatric to adult care in patients with inborn errors of metabolism

Due to improved treatments and prognosis, many diagnosed with inborn errors of metabolism during childhood reach adulthood. An article published in *Medicina Clínica (English Edition)* provides recommendations on how to transition from paediatric to adult care for these individuals. After a thorough bibliographic survey, the authors provided a list of obstacles emanating from the patient, family, health

services or hospital infrastructure that may interfere with the transition process. They then provide recommendations for the transition process in patients with inborn errors of metabolism:

- "Carry out the transition between the age of 16–18, at a time of disease stability and taking into account the level of development of the patient.
- Carefully assess situations of palliative care before deciding to make the transition.
- Develop a transition plan, flexible and individualized, with the patient and family.
- Provide the patient and his/her family with information about IEM and the treatment plan.
- Find an adult team, and a coordinating professional (preferably an internist) who can coordinate, together with the paediatrician, the process and who can ensure continuity of care for IEM patients.
- Encourage the creation of specific attention protocols for these diseases and the formation of multidisciplinary teams specialized in IEM. "

To read the abstract (US$31.50): http://www.sciencedirect.com/science/article/pii/S2387020616307811

Rare Disease Northeast USA Summer Family Camp 2017

NORD is once again partnering with The Hole in the Wall Gang Camp on a rare disease summer family camp in Connecticut. The camp provides a special opportunity for children and families impacted by rare diseases to join together for a weekend of pure fun - free of charge. Camp is open to 25 families who are located in the Northeast and it will take place June 1-4, in Ashford, CT. Apply here

Exceptional Parent magazine's 2017 Annual Resource Guide (USA only)

eParent Special Needs Resource Directory provides a comprehensive guide for US-based parents and families of children with chronic health issues, disabilities and special needs, as well as to physicians, allied health care professionals and educational professionals who are involved in their care and development.

The directory is designed to assist in the search for helpful information and resources for every situation. This year it has an expanded military section.

Available here and here

TELL US!!

If you have a child or loved one that is facing surgery or dealing with an illness, please notify us at info@ismrd.org and we will send them a greeting in our newsletter.

If you are a family that has lost a loved one to any of the diseases ISMRD covers, please let us know so that we can acknowledge their passing in the newsletter.

We would also love to print your family story, so please send it to us.

ISMRD

ISMRD'S Sunshine Care Committee

ISMRD has a group of parent volunteers called the **"Sunshine Committee"**. Our purpose is to coordinate support for families in need. The type of support varies on the circumstance -- from birthday and weddings, an illness or death in the family, or a family experiencing surgery or a medical crisis. In any case, we provide a little "sunshine" for the family by providing flowers, encouraging messages via email, cards or a phone call -- whatever we think the family would find most helpful. In order to help others, our group relies on the support of all families because, in essence, we are all part of the ISMRD "Sunshine Committee".

If you are in need of assistance or know someone in our Penguin community who is in need, **please contact Susan Kester**. She will coordinate with the appropriate parties to determine how we can best help.

ISMRD gratefully acknowledges

- Andrea Gates
- Baumeyer Family
- Beckie Leah
- Carolyn Paisley-Dew
- Catholic Daughters of the Americas
- ClearView Healthcare Partners
- David Dollins
- Family of Joyce Blesi Knaus
- Glen Howard
- Julie Kahn
- L B Boughfman
- Mark Stark
- Mary Bohannan
- Mary Kimmet
- Martin Woolley
- Maurice and Betty Daniken
- Nancy Kourik
- Pam & David Tobey
- Phillip Fields
- Robin Montgomery
- Ultragenyx Pharmaceutical Inc
- Wagner Foundation

for their very generous donations

ISMRD would like to send condolences and make a special thank you to the family of Joyce Blesi Knaus.

Joyce was the aunt of ISMRD President Jackie James and her husband Bret. Joyce passed away recently and her family asked that people donate money to the ISMRD in Joyce's memory, in lieu of sending flowers.

Thank you to Joyce's family, and those that made a donation to the ISMRD in Joyce's honour.