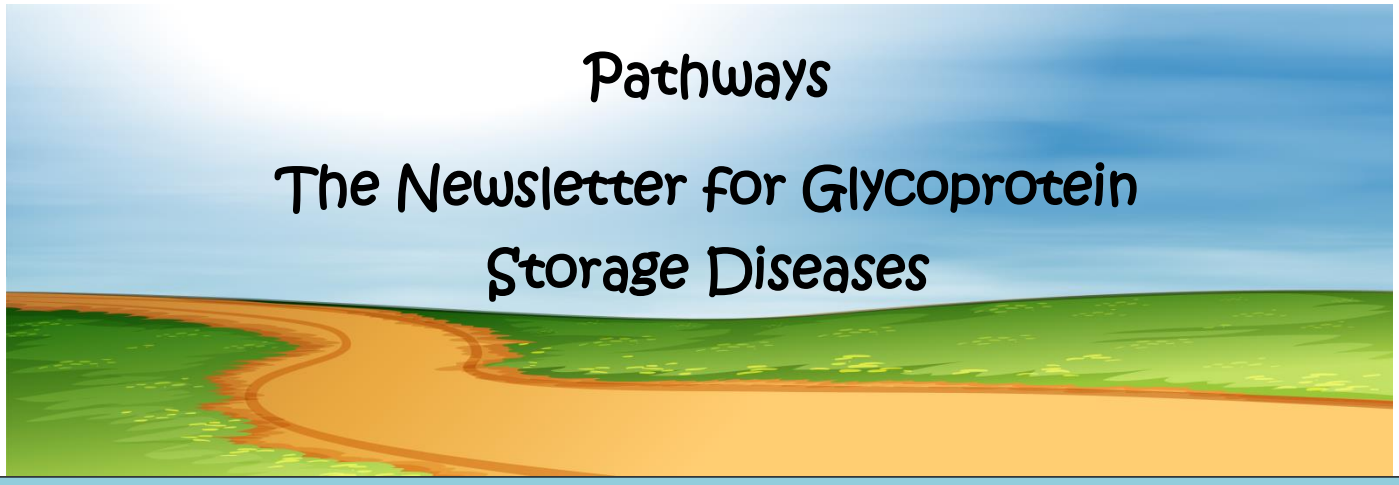

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Our Mission and Vision

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 and 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 Diseases and Sialidosis

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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

Contact Us

The International Advocates for Glycoprotein Storage Diseases
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USA
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From the President's Desk

By Mark Stark
President, ISMRD

Welcome to the ISMRD July newsletter!

As announced in the last newsletter, the ISRMD board has begun serious work to plan for the next combined scientific and family conference scheduled for 2015. A lot of work has been done by our board members and our professional advisory board to raise funds and identify topics and speakers, and I am very happy to report that the Fourth International Conference on Glycoproteinoses is scheduled for July 23rd to 26th, 2015. More details about the conference are described in the newsletter below. Of course, there is still a lot of work to do, including fundraising to ensure more researchers and families can attend the conference. We very much appreciate your help in supporting this effort, and this newsletter includes some easy ways that you can help contribute to the conference. The conference will further our mission at ISRMD to help all those affected by rare diseases by building partnerships with medicine, science and industry in methods to detect and cure these diseases, and to provide a global network of support and information.

Another aspect of the ISMRD mission is enabling research for these rare diseases. I am also pleased to announce a partnership grant with the MPS Society to provide research funding for Mucolipidosis II/III. More details about this research grant are attached in the newsletter. I very much appreciate the generosity of our donors who have made it possible for ISRMD to enable this research, and I would also like to thank the MPS Society for their donation and leadership in selecting a deserving project.

This newsletter also provides information to families who may not be in regular contact with ISRMD; providing news about activities and upcoming events that are important to everyone concerned with curing these debilitating disorders. This edition of the newsletter continues a focus on information about resources that are available for caregivers and families. There is a summary of interesting research on the frequency of metabolic disorders by region, and a list of upcoming conferences around the world. Last, but not least, I would like to personally welcome the 12 new families that have joined the ISMRD family.

I would like to thank everyone who has already contributed this year to ISRMD; both financially and with your time. We are continuing our fundraising campaign for the 2015 combined scientific and family conference on glycoprotein diseases. If you would like to help – financially or otherwise - please contact any of our board members, and we will be happy to find ways you can participate.

Mark Stark
President
ISMRD

SAVE THE DATE AND JOIN US FOR THE 4th ISMRD INTERNATIONAL FAMILY AND SCIENTIFIC CONFERENCE!

The ISMRD Board of Directors is honoured and excited to once again sponsor a combined Scientific and Family conference for Glycoprotein Storage Disease in July 2015.

The conference:

Glycoproteinoses: Fourth International Conference on Advances in Pathogenesis and Therapy

Date: 23rd – 26th July 2015, St Louis, Missouri, USA

Venue To be advised

We are very pleased to announce Dr. Stuart Kornfeld *(see Dr Kornfeld's Bio below*) has agreed to be the primary investigator for the scientific conference. Dr. Kornfeld's vision for the conference is: "The Fourth International Conference on the Glycoproteinoses will bring together leading investigators from around the world to discuss the latest advances in understanding the pathophysiology of these rare disorders and the status of the development of new therapies. The intent is to stimulate an interchange of ideas and develop collaborations among investigators with different approaches and expertise. Another goal is to increase the awareness of these underserved forms of lysosomal diseases among new investigators, post-doctorate fellows and graduate students. The conference is designed to foster interactions between the investigators and patients/affected families."

One of the primary objectives of the conference is to provide more information to families and give them the chance to meet with doctors and researchers for these rare diseases. This was a very successful part of the 2012 conference, and ISRMD is again sponsoring a family program. Dr. Barbara Burton (*see Dr Burtons Bio below*) has agreed to lead this program.

Our goal is to have the largest possible participation of investigators and families. To do this, ISMRD has launched a fundraising campaign, and we have already received significant corporate sponsorship. However, every donation we can receive for this conference will help another family or young investigator attend this conference. If you are interested in donating to the conference please **click**

We look forward to welcoming you all to St Louis in July 2015. Please keep your eye on our website as we start to develop the conference website.

We are proud and honoured to also announce our Scientific Chair Professor Stuart Kornfeld and Dr Barbara Burton who will be helping to put together the family program.

Stuart Kornfeld, MD

Professor
Department of Medicine
Hematology Division
Department of Biochemistry & Molecular Biophysics

His clinical interests are in General Hematology and his research interests are Intracellular Protein Trafficking Lysosomes, Glycoproteins

Barbara Burton, MD

Professor
Pediatrics-Genetics, Birth Defects and Metabolism
Northwestern University Feinberg School of Medicine

Her clinical interests are in PKU and other metabolic disorders, lysosomal storage disorders, neurogenetic disorders, connective tissue disorders, MPS disorders, Morquio syndrome
Description of Interests

About St. Louis is an independent city[7] and a major United States port on the eastern line of the state of Missouri. At the 2010 census, the population was 319,294, and a 2013 estimate put the population at 318,416,[6] making it the 58th-largest U.S. city in 2013. The metropolitan St. Louis area, known as Greater St. Louis (CSA), is among the 20 largest metropolitan areas in the United States with a population of 2,900,605.

The city of St. Louis was founded in 1764 by Pierre Laclède and Auguste Chouteau, and named for Louis IX of France. After the Louisiana Purchase, it became a major port on the Mississippi River; in the late 19th century, it became the fourth-largest city in the United States. It seceded from St. Louis County in March 1877, allowing it to become an independent city and limiting its political boundaries. In 1904, it hosted the Louisiana Purchase Exposition and the 1904 Summer Olympics. The city's population peaked in 1950, then began a long decline that continues in the 21st century. Immigration has increased, and it is the center of the largest Bosnian population in the world outside their homeland.

Some Highlights of St. Louis

The Gateway Arch, City garden, Butterfly House, Saint Louis Art Museum, Saint Louis Zoo, Missouri History Museum, Saint Louis Science Center, Anheuser-Busch Brewery, The City Museum, Grant's Farm, The Museum of Transportation

ISMRD supports research for Mucolipidosis

In 2013, ISMRD and the National MPS Society offered a Partnership Grant for Mucolipidosis II/III. The grant is $20,000 for each year of the two years. Following a global request for proposals which were reviewed by a committee comprised of members of our Scientific Advisory Committees, the grant was awarded December 2013 to Dr. Heather Flanagan-Steet at the Complex Carbohydrate Research Center at the University of Georgia. ISMRD is grateful to the National MPS Society for helping to, ensure the success of this endeavor.

ISMRD wants to thank all the families who have raised funds which has allowed us to undertake this research project. Below is the abstract for the project we are supporting which will be presented at the 2015 Conference.

"Investigating the role of cathepsin proteases in ML-II cardiac pathology"

Heart valve defects represent a life threatening but poorly understood symptom of ML disease. Recent work in our ML II zebrafish model has provided new information on why the valves don't form or function properly. Our earlier work on cartilage defects in this model identified the enzyme, cathepsin K, as a central player in the disease process. Inhibition of cathepsin K in the ML II background resulted in improved cartilage development, suggesting a new therapeutic strategy for ML disease. Since the development of heart valves and cartilage share many common features, it is likely that cathepsin K also contributes to ML heart valve disease. We propose to use inhibitors of cathepsin K (and another related enzyme cathepsin L) to ask whether they reverse the heart valve defects present in ML II zebrafish. At least one cathepsin K inhibitor, odanacatib, recently passed Phase III clinical trials for the treatment of osteoporosis and is expected to be available in 2014. Our proposed work may uncover a new opportunity to treat ML valve disease with these inhibitors.

ISMRD FUNDRAISING

ISMRD is currently raising funds through GoFundMe. We hope to be able to raise $70,000 to help support the Natural History Study, thus ensuring that new families are able to participate in the study.

To make this fundraiser work, we are asking you our families and those searching our website to share the link via your social media links such as Facebook and Twitter.

Please visit our GoFundMe page to make a donation and to meet some of our very special people.
http://www.gofundme.com/5rpjhw

ISMRD has registered with AmazonSmile, which is a simple and automatic way for you to support ISMRD every time you shop, at no cost to you. When you shop at smile.amazon.com, you'll find the exact same low prices, vast selection and convenient shopping experience as Amazon.com, with the added bonus that Amazon will donate a portion of the purchase price to ISMRD. Go to http://smile.amazon.com

ISMRD Calendars –This was such a successful event. ISMRD thanks all the families who submitted their children's photos and sold them as Christmas gifts. We will be preparing the 2015 Calendar ready for Christmas sales again.

Help required: ISMRD is looking for a family member who would be happy to gather photos and put the calendar together and get it ready for Christmas sales. Please contact Susan Kester info@ismrd.org if you would like to take this task on.

What's happening around the world?

12th European Conference on Rare Diseases: Living with a Rare Disease

4–6 July, 2014, Spala, Poland

The objective of the Conference is to introduce the multifaceted nature of rare diseases. The Conference, seminars and training on rare diseases are there to make everyone aware that coordinated actions will allow to improve the quality of life of patients with MPS and rare diseases in Poland and all around the world. **Go to** http://chorobyrzadkie.pl/materialy/konf.2014.zap.en.pdf

3rd Nordic Conference on Rare Diseases

4-5 September, Helsinki, Finland

NCRD 2014 will be the 3rd Nordic conference focusing on the rare diseases and current topics related to them. NCRD 2014 introduces for example the national plans or strategies for rare diseases made in Nordic countries, touching the implementation of the plans and the experiences gained so far. The conference offers an excellent opportunity to network and to support the exchange of best practices throughout the Nordic countries. Joint action helps patients and professionals share expertise and information across borders. **Go to** http://www.harvinaiset.fi/Helsinki2014

Newborn Screening in China

PerkinElmer, a Massachusetts-based company with neonatal and Newborn Screening (NBS) technologies, has announced a collaboration with China's National Health and Family Planning Commission to develop and implement a 3-year NBS screening training program focused on early detection of life-threatening diseases in rural areas of China. Last year, PerkinElmer screened more than nine million newborns in China.

Some of the key features of the NBS Program are to train more than 3,000 doctors, clinicians, and laboratory technicians in 600 rural counties in :

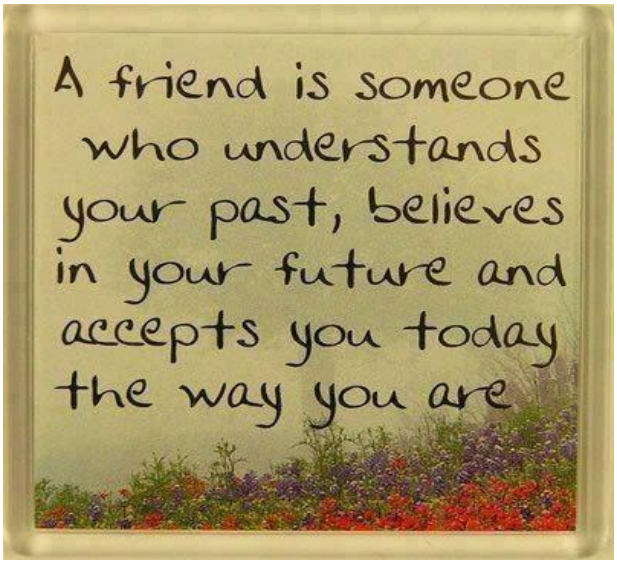
- Sample collection
- Clinical diagnostics and treatment
- Project management.

Varying incidence of inherited metabolic disorders among countries

A September 2013 article, titled "Newborn Screening for inherited metabolic disorders; news and view", in the *Journal of Research in Medical* Sciences, presents results from expanded newborn screening programs using tandem mass spectrometry have revealed that the incidence of inherited metabolic disorders varies considerably among countries. For example the combined incidence of amino acidemias, organic acidemias, and fatty acid oxidation defects are as follows :

Country	Incidence
Qatar*	1 in 1,327
Korea	1 in 2,000
Germany	1 in 2,517
North America	1 in 4,000
Hong Kong	1 in 4,122
Denmark	1 in 4,900
Mainland China	1 in 5,800
Taiwan	1 in 5,882
Australia	1 in 6,369
Japan	1 in 9,330

*Qatar has a high rate of consanguinity to read more
http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3872591/



Support Resources

Global Genes Project

Congratulations to the Courageous Parents Network on launching their website! "Coping Resources for Parents Caring for Children with Life-Limiting Illness: Shining Light on the Dark Places"

To read more: http://courageousparentsnetwork.org/

The Caregiver Space

TheCaregiverSpace.org is a free social network that allows caregivers to share their experiences, find critical resources, cope with stress and effectively combat the isolation and exhaustion of providing care for someone they love.

A safe haven for family caregivers and professional carers alike, The Caregiver Space offers 24/7 access for members to freely discuss respite care, caregiver burnout and all the difficult emotions and stressors that accompany ongoing support to a loved one.

To read more: http://thecaregiverspace.org/

Free Rare Diseases E-Book - Rare Diseases – Diagnosis, Therapies and Hope

This book focuses on all rare disease audiences and includes:

- Ten case studies of amazing discoveries from rare disease families
- Many interviews with top doctors, researchers and genomics experts
- A Rare Disease Parent's Toolkit with recommendations from parents and others
- Demystifying Genomics, which simply explains the latest revolution in medicine with plenty of information on how the sequencing process works.

To read more: http://griefhelp.org/the-road-to-recovery/

New Online Tool For US Patients Searching For Medical Experts

CurityMD created by start-up SpecSo Health is a new tool that uses data, statistics, and technology to connect patients with rare diseases to the appropriate information and care that is required. CurityMD is "an online platform that helps measure expertise in rare and complex conditions to help improve how pateints and qualified specialists connect". CurityMD is a search engine for rare diseases. With CurityMD one can:

- Find the most experienced care centers and experts, nearby or anywhere in the United States
- Find Important clinical trials, research and new treatments
- Find hospitals looking for patients with rare and complex conditions.

To read more: www.spesohealth.com

ISMRD warmly welcomes the following people to our family

- Kimmy Macedo-Galache and her daughter Brielle who has ML II. They live in the USA
- Gavin and Estelle Blake whose daughter Gabriella has ML III. They live in South Africa
- Patty Covarrubias Coronado
- Joyce Amick
- Avinash Pudrod, India
- Shantel Marie and her daughter Shyanne
- Kataelynn Dickey and her daughter Emmerson Jayne who has ML II. They live in the USA
- Kristi Johansen Santeramo and her 15 year daughter Glynnis who has Sialidosis. They lives in the USA
- Anthony Donachie
- Marry Van der Plas whose child has ML II. They live in the Netherlands
- Brooke Randa Malesberger, whose daughter has ML II
- Heather Trev Scott who lives in England and has ML III

May the stars carry your sadness away,
May the flowers fill your heart with beauty,
May hope forever wipe away your tears,
And, above all, may silence make you strong

Sadly we mourn the loss of:

- Antonn Peach who lived in New Zealand and had Sialidosis. He passed away in March 2014
- Gracie Sims who lived in England and had ML II. She passed away on 29th May aged 18 months.
- Jonalin Coley from the USA who had ML II. She passed away on 19th June 2015 aged 11 years

If you know of anyone who has recently been ill or had surgery or is about to have surgery, please tell us at info@ismrd.org

Some of our Penguin children and young adults have recently been in hospital, had surgery or are awaiting surgery

Your Penguin family are thinking of you and praying for a good outcome

- Gabby Blake, ML III, hip replacements
- Shyanne, ML II
- Sergio Cardona-Cardenas, ML II/III
- Bianca Trestianu, ML II
- Taryn Murphy, Alpha Mannosidosis
- Ariel Buchanan, ML lll, hand surgery
- Timmy Buchanan, ML lll, ear tubes and adenoidectomy
- Jenny Klein ML lll, hip surgery
- Katelyn Smith, ML lll, hip replacements

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 at info@ismrd.org** She will coordinate with the appropriate parties to determine how we can best help.

ISMRD gratefully acknowledges the following people for their very generous donations.

Without this kind of support we would not be able to carry out our mission and vision for ISMRD.

- Ana Martinez
- Juanita Van Dam
- Han Petter Franck-Nielsen
- Hilde Skulstad
- Carolyn Paisley-Dew
- Canberra Public Servants Union, Canberra Australia
- Carolina Verde
- Ash Van Dijk
- Stephania Semova
- Trish Dennis
- Interior Office Solutions
- Andrea Porch, Coomera River State School, Queensland Australia in honour of Damian and Jesse-Rose Van Dam