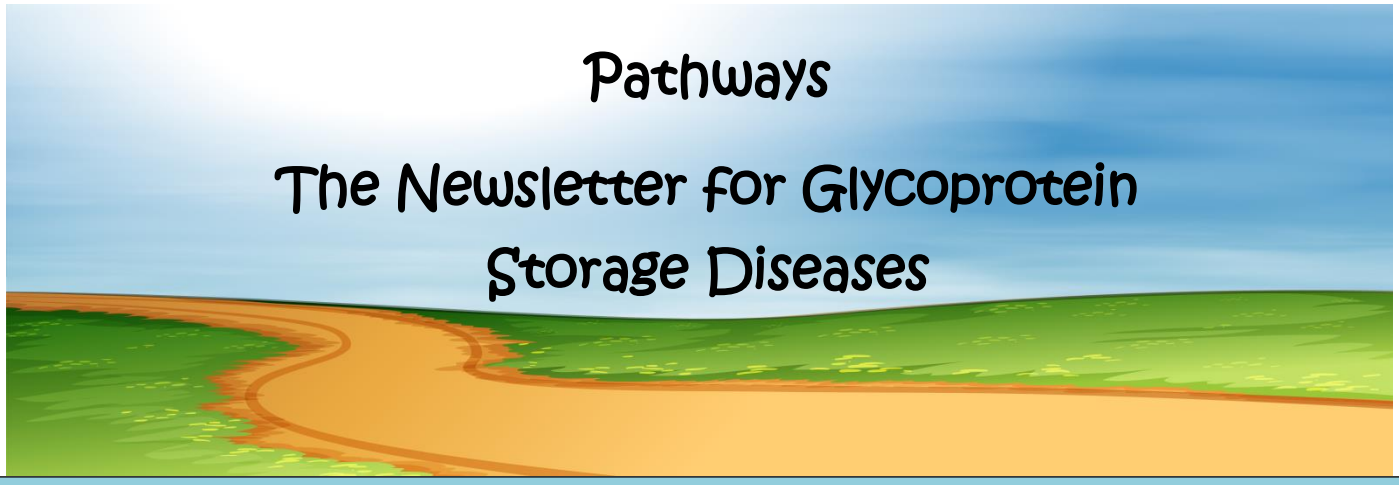


ISMRD, a 501 © not-for-profit organization, FEIN 53-2164838 | website www.ismrd.org

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 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

In this Issue

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
20880 Canyon View Drive
Saratoga
CA 95070
USA
email: info@ismrd.org

From the President's Desk

By Mark Stark
President, ISMRD

Welcome to the ISMRD December newsletter. I hope that you are lucky enough to be spending your holidays with friends and family! In this happy season, it is important that we remember those families that are dealing with the loss of a loved one. I know I speak for everyone on the board of ISRMD when I say our thoughts and prayers go out to anyone who has lost a loved one this year. This has hit especially close to home this year for ISRMD. One of our board members, Pam Tobey, lost her daughter Autumn last month. I am also sorry to inform you that another of our board members, Andrea Gates, is suffering from a severe illness. I hope you will include Pam and Andrea in your thoughts and prayers.

I would like to thank everyone who has sent remembrances in honor of those children who passed this year. We plan to use funds donated to help bring more families to the upcoming conference in St Louis, and to fund research to better understand and cure our children's disorders. Thank you to everyone who has donated in any way this year, with a special thank you to the Kimmet and Woolley families who contribute every month.

Registrations open soon for the Fourth International Conference on Glycoproteinoses, which is scheduled for July 23 - 26, 2015 in St Louis, Missouri. We very much hope that we will see you there. This newsletter includes some easy and fun ways that you can help raise funds for your conference accommodation or for the conference in general, by selling special bracelets and buying the 2015 ISRMD calendar.

I would very much like to thank all those who helped put together and distribute the 2014 and 2015 calendars, and all those who helped with the 2014 Rare Disease Day mailings.

Information about the 2015 Rare Disease Day is available in the newsletter.

We are also seeking new Board members to assist with fundraising and communications.

The newsletter this month is packed with interesting and informative articles for your holiday reading. There is a touching story about a UK family with two children with Fucosidosis. There are links to the latest news stories about rare disease research and therapies, and information about support resources.

I hope that you and your loved ones have the opportunity to share the love, joy, and peace of this season. Enjoy the time, and have a great start to 2015!

Thank you
Mark Stark
President
ISMRD

Registrations for the

Fourth International Conference on Glycoproteinoses

open on

1 January 2015

Go to:

http://www.ismrd.org/fourth_international_conference/registration

Early Bird registration closes on 30 March 2015

ISMRD Is Seeking New Board Members

Would you like to be involved in the management and future directions of ISMRD?

We currently have vacancies on the Board in the following areas:

Fundraising. This person would have experience in seeking corporate funding and in grant-writing. They would work with our existing Fundraising Team to organise fundraisers and with our Research Committee to investigate funding for the continuation of the Natural History Study.

Communications: This person would have a wide knowledge of social media, be able to write press releases and help us raise the profile of ISMRD around the world. They would help the existing Communications Team to develop ISMRD's International Rare Disease Day profile and events.

All ISMRD Board roles are voluntary. If you think you would enjoy one of the above roles, and have the relevant experience, please send a one-page letter outlining your skills and experience to info@ismrd.org

Raise money for your conference accommodation costs -

new bracelet colours now available

Our beautiful fundraiser bracelets are now available in red, green, purple and pink, as well as the original blue

The bracelets sell for USD$20. For every bracelet you sell, ISMRD will put aside USD$10 in your name to cover some or all of your accommodation costs.

If you would like to purchase some bracelets to sell to your friends and family, please send an e-mail to **info@ismrd.org**.

If you are not attending the conference, you can sell bracelets and nominate a family that is attending to receive the $10 towards their accommodation costs.

gofundme

Don't forget the ISMRD gofundme page, which has so far raised US$2,660. Every little bit helps. If you would like to donate, go to http://www.gofundme.com/5rpjhw

Amazon Smile

If you shop at Amazon Smile, a portion of the purchase price will be donated to theo ISMRD, at no cost to you. **Y**ou'll find the exact same low prices, vast selection and convenient shopping experience as Amazon.com. Go to http://smile.amazon.com

Shop at AmazonSmile
and Amazon will make a donation to:

Get started

amazonsmile

ISMRD 2015 Calendar now available

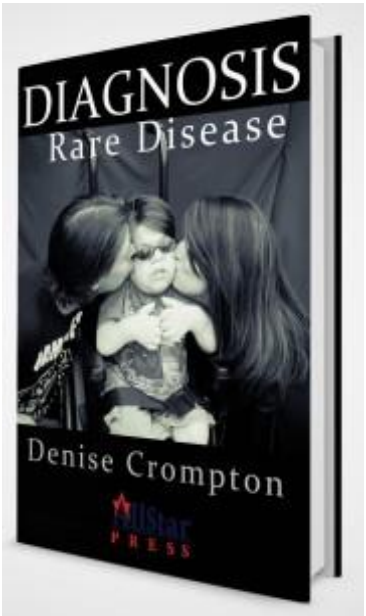
$14 each

The ISMRD 2015 calendar is now available. It costs USD$14, and features more of our kids than last year.

See a preview at http://www.ismrd.org/…/pdf_fi…/0020/45326/2015_ISMRD_cal.pdf.

Order your calendars from Susan Brennan Kester or at http://www.ismrd.org/fundraisers.

Proceeds will go to the 2015 International Scientific/Family Conference being held in St. Louis. We hope that you will embrace this fundraiser and help us raise funds for the conference where many families will be meeting other families with the same condition for the very first time.



DIAGNOSIS: RARE DISEASE
by ISMRD member Denise Crompton
A Big Success

AllStar Press announces: "Within 24 hours of being released in September, Crompton's book hit the #1 spot for *paid books* in the Genetics category of Amazon.

You will absolutely love this book or your money back."

Professor Jules Leroy says: " The book is to become a classic in the still little-known world of very rare or orphan hereditary disorders."

You can order your copy in paperback from AllStar Press, Amazon Smile and Barnes & Noble. It is also available electronically in Amazon Smile and in all digital ebook formats through Smashwords.

Please note that the author's royalties are going toward research.

Family Story

Stephen and Lauren Bell are 22 and 16
They live in the United Kingdom and have Fucosidosis

Their mother, Linda Bell, tells their story

You never know what the future holds, sometimes it's just as well.

I had never planned or had a great desire to have children, it just wasn't something that was on my wish list.

All that changed when aged 34, I met my husband Graham, marrying a year later, and then the concept of us becoming a family started to appeal. Pregnant with my first child at 36, I had an amniocentesis test and was reassured that all was fine.

Stephen was born four weeks early, but otherwise all ok weighing 6lb-10ozs, a beautiful, welcomed and much loved little boy. He reached his baby milestones at the right time, although his speech was slow. Some initial concerns started to be raised once he started school because he was falling behind his peers with a widening gap. I think I was in denial that my perfect boy could have any issues and would challenge and make excuses if any concerns were raised. The only physical symptoms evident were that Stephen could be a bit clumsy and has a small red mark behind one ear which I thought of as a birthmark, it always seemed to have been there.

When Stephen was six, he was joined by his sister Lauren, who again started off reaching her baby milestones at the correct time but from being a toddler some concerns started to be raised about her development. I was in denial. I don't think I could see or accept that there was anything wrong. Lauren looked very similar in appearance to her brother but at around 18 months a small red circle of dots appeared on her bottom which I assumed was a sort of rash or birthmark. It slowly grew bigger and she had a sort of red mottling under her skin.

When Stephen was at junior school, he continued to fall behind his peers and the school suggested a statement of special needs to provide additional support. At one point a concern was raised about the red mark behind his ear, whether this could be evidence of any abuse. That was very frightening, the thought that a doctor could be thinking that way and could have resulted in significant consequences. That issue was not pursued when the red mark was still there but unchanged a week later. Apparently some children are abused by pulling their ears which causes bruising which would start off red but change colour as the bruise healed.

Anyway a paediatric doctor, doing a routine check posed the question: "could Stephen's delayed development with the red mark and skin mottling and Lauren's red skin marks be linked to something?". We were sent to Alder Hey Children's hospital for tests including genetic tests. It was at this point that we first heard the name Fucosidosis, the condition which both children had. A very rare condition which made my special children even more special in some ways.

I did the worst thing possible and went home to Google the condition and cried myself to sleep after reading about Fucosidosis and the outlook and implications. Frightening descriptions about shortened life expectancy, perhaps not reaching age 10, although Stephen was already older than this.

Having a label at least explained why Stephen sometimes did not remember some of the topics at school and had to have things repeated. He wasn't just being lazy.

Lauren's red marks had become more noticeable over time and now covered a larger area on her bottom and legs, they could be unsightly but did not hurt or cause her any issues but could bleed heavily if knocked. We now knew they were called Angiokeratomas. They could be quite raised at times, were not static and could disappear and reappear elsewhere. We later learnt about how and why this occurred.

The diagnosis started review checkups with Dr Ed Wraith, a lovely specialist at Alder Hey hospital who provided explanations and answered questions to the best of his ability at the 6-monthly review meetings. It was hard to accept that there was no cure, medication or treatment although symptoms could be treated. We often had to explain the condition to others, teachers, doctors, dentists etc, as no one had ever heard of it.

Both children continued to receive support at school but the same condition has not affected them in the same way.

Stephen was very prone to colds and ear infections as a child although he has outgrown much of this. He is now 22 years old. He managed with support to get seven exam qualifications at High School. He suffers quite badly from a speech impediment and is clumsy and awkward in his movement. His back will bend sideways if he has to walk a distance or stand for a long period. This is down to some issue with his spinal column. He has a heart murmur which is monitored but has not required any treatment to date. He can be affected by upset stomach problems and Irritable Bowel Syndrome type problems on occasions. He has however been able to undertake some work experience, voluntary work and even some ongoing paid employment working as a sorter for the Royal Mail. He can travel independently in the local area by bus and has done so since he was a teenager but does not drive and still lives at home. He could possibly at some time live independently with support, although not at this time.

Lauren on the other hand is much more vulnerable and much less independent. She is now 16. She is physically healthy, however she has more red Angiokeratomas on her body and her condition has resulted in mental issues. About two years ago, she suddenly deteriorated and lost touch with reality, talking to herself and going into her own world, constantly on the go day and night and not sleeping. After tests, there appeared to be no other additional problems but a change in the condition. The hospital described this as a chronic brain disorder. After about a year she did improve somewhat but she has recently

deteriorated back into her own world and this is where she is at the moment. We have currently lost the personality of our daughter, no longer doing the things she enjoyed even a couple of months ago, dancing to her music, singing along to and watching soap operas. She appears happy, but in her own world. We are just hoping that over time, like the previous episode, she comes through and out from her prevailing state.

Lauren is attending special needs 6th Form College but at this time is not independent in any way even to the point of having to check and oversee her as she eats, bathes, dresses herself. Graham, her father is now her full time carer and I am about to leave work and retire early to also care for Lauren.

Both our children are of course very much loved but have changed our lives in ways we could not have imagined, and of course we worry about the future for both them and us. At times their condition is hard to accept, you get on with dealing with how things are but from time to time it feels unjust that this isn't fair and the condition is cruel for they are so different from their peers.

However there are no guarantees that life should be fair and things could be worse.

Competitions

EveryLife Foundation For Rare Diseases: Annual Rare Artist Contest

The EveryLife Foundation for Rare Diseases is holding its 6th Annual Rare Artist Contest. The deadline for entries is 16 January 2015, and winners will be announced by February 15, 2015.

The **EveryLife Art Contest** was established in 2010 for artists affected by a rare disease with the purpose of raising awareness of rare diseases and to encourage the telling of their stories using art. The mission of the contest is:

" … to showcase the artwork and the Artist, in order to raise awareness about rare diseases and showcase our vibrant community. Art is the expressive medium used to capture the trials and triumphs of our human existence. For rare disease patients, life can be a day-to-day struggle wrought by a devastating disease and unanswerable questions. Art as an expression of these challenges in life and in living each day, can be a particularly powerful medium to guide all of us, in our struggles and successes. Artists affected by rare disease communicate their pain, frustration, optimism and joy, and through their work we can learn more about ourselves … ".

To read more, go to http://www.rareartist.org/2014-art-contest/

What's happening around the world?

Saturday 28 February 2015 will be the eighth international Rare Disease Day. The official theme for Rare Disease Day 2015 is "Living With a Rare Disease." The slogan for 2015 is: "Day by Day, Hand in Hand."

The main objective of Rare Disease Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives.

The campaign targets primarily the general public and also seeks to raise awareness amongst policy makers, public authorities, industry representatives, researchers, health professionals and anyone who has a genuine interest in rare diseases.

Since Rare Disease Day was first launched by EURORDIS, the European Organisation for Rare Diseases, and its Council of National Alliances in 2008, thousands of events have taken place throughout the world reaching hundreds of thousands of people and resulting in a great deal of media coverage.

The political momentum resulting from Rare Disease Day also serves advocacy purposes. It has notably contributed to the advancement of national plans and policies for rare diseases in a number of countries.

Even though the campaign started as a European event, it has progressively become a world phenomenon, with the USA joining in 2009, and participation in a record-breaking 84 countries around the world in 2014. Some countries have decided to raise rare disease awareness further, for example, Spain declared 2013 as the National Year for Rare Diseases.

There are many ways you can participate on the day. To get involved, go to http://www.rarediseaseday.org/article/get-involved

Read more: http://www.rarediseaseday.org/ http://www.rarediseaseday.us

US Survey Shows New Parents Interested In Newborn Screening

514 parents at Brigham and Women's Hospital were surveyed within 48 hours of their child's birth to establish their views on genomic newborn screening.

The following results were observed:

- 18.1 % of parents extremely interested in testing for their newborn
- 28.0 % of parents very interested in testing for their newborn
- 36.6 % of parents somewhat interested in testing for their newborn
- 10.9 % of parents a little interested in testing for their newborn
- 6.4 % of parents not at all interested in testing for their newborn.

The survey results were similar regardless of parents' age, gender, race, ethnicity, and other factors. However, if a parent "experienced concerns about the health of their newborn, they were less likely to be interested in genomic testing."

Read more: **Newborn Screening Survey**

The Five Most Expensive Drugs In The World Are All Orphan Drugs

Orphan drugs Acthar Gel, Cinryze, Kalydeco, Naglazyme and Soliris are the five most expensive drugs in the world, in ascending order of price. While these drugs have life-altering properties for rare-disease patients, it is possible that, in the US, the exorbitant price could influence insurers to opt out of offering these drugs in their approved drugs list. This could be disastrous for both the pharmaceutical companies and patients.

Read more: **Five Most Expensive Drugs**

Research Consortia Will Study More Than 200 Rare Diseases

The US National Institutes of Health has announced the expansion of its Rare Diseases Clinical Research Network (RDCRN). Six new and 16 continuing consortia as well as the RDCRN Data Management and Coordinating Center will be funded. More than 90 patient organizations will work with researchers to study more than 200 rare diseases.

Read more: **Research Consortia**

Scottish Funding For Rare Disease Drugs

Scottish Health Secretary Alex Neil has announced the expansion of a special fund giving patients in Scotland access to expensive drugs.

A fund of £40m will be available this year and next year for new drugs which are not normally paid for by the NHS.

The New Medicines Fund doubles the amount of money previously available.

Read more: **Scottish Rare Disease Drugs Funding**

Swiss National Plan for Rare Diseases

The Federal Council of Switzerland has approved the "National Concept on Rare Diseases", which is an equivalent of the countries' national plan for rare diseases. The plan proposes 19 measures, including the establishment of reference centers designed to ensure a quick and safe diagnosis as well as effective treatment. The implementation of this plan is scheduled for the spring of 2015.

Read more: Swiss National Concept on Rare Diseases

Read the National Concept Rare Diseases in French
Read the National Concept Rare Diseases in German

Dental Center For Children With Special Needs In Lebanon

Lebanon has opened a dental centre for children with disabilities. The centre provides care that is equivalent to that offered by other international reference centres for oral manifestations of rare diseases. The collaborative team consists of a physician anaesthesiologist, an autism specialist, a paediatrician, a nutritionist, a specialist in biomolecular research, a multidisciplinary dental team and search platform in molecular biology from the Faculty of Science at the Lebanese University. A partnership has also been established with the National Reference Centre for Dental Manifestations of Rare Diseases, University Hospital of Strasbourg.
For further information contact **Prof. Elia Sfeir**

See Penguins Dressed for Christmas in Korea

Go to: Christmas Penguins in Korea

Thank you to Lisa Patriquin for

Carer Resources

Mothers of children with rare disease go online to combat their sorrow

The *Journal of Pediatric Nursing* has published a study analysing how families affected by rare diseases use online communications to manage the extremely difficult and emotional journey. The authors note that the isolating nature of rare disease and the unfamiliarity of many healthcare providers complicate the management of the condition and psychosocial experiences make mothers, who are more likely to search the internet for health related information, use online support groups. In this study the authors interviewed mothers of children with Alagille syndrome to understand how they used online health communications to manage their chronic sorrow. They found that not only did online communication impact the emotional state of these mothers but also provided them knowledge to manage the disease and educate others, including healthcare providers. The authors report that the mothers appreciated being part of the online community and meeting others in similar circumstances but did acknowledge that it sometimes triggered unpleasant feelings or chronic sorrow and sometimes faced an information overload.

Read the abstract of the study: **Online Effects for Sorrow**

Free Cake for US children

Icing Smiles is a nonprofit organization that provides custom celebration cakes and other treats to families in the US impacted by the critical illness of a child. To apply for a free cake, go to Icing Smiles application form

Thank you to Danielle Z for this tip.

ISMRD warmly welcomes

Sally Thornton-Helmer

to our family

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

- **Autumn Tobey**
  who passed away on 22 November 2014, aged 38.
  Autumn had Mucolipidosis lll

- **Wynnie Johnson**
  who passed away on 2 December 2014, aged six.
  Wynnie had Mucolipidosis ll

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

- Sergio Cardenas, ML II/III
- Gabby Blake, ML lll
- Jake Glover, Fucosidosis
- Bobbie Gross's daughter, Alpha Mannosidosis
- Heather Scott, ML lll
- Saffy Woolley, Alpha Mannosidosis

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 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.

- Mary Bohannan
- Bill & Alice Bowen
- Alicia Chessir
- Bob & Denise Crompton
- Doris Dooly
- Shelby & Jude Fenter
- Larry & Elizabeth Godfrey
- Ruth Hatwig
- Bill & Dona Holzman
- Carol Holzman
- Margaret Kelly
- Mary Kimmet
- Jackie James
- Les Jarrard
- Roy & Dianne Jones
- Susan Kester
- Edel Lougheed
- Lorene Mantooth
- Tricia May
- Parker Meador
- Anita Muonio
- Melissa Neissl
- Tim & Gena O'Neal
- Nicole Potts
- Diane Sanders
- Stephania Semova
- Edward & Carole Shaw
- Margaret Sisti
- Charlotte Taylor
- Norma Vaugn
- Katrina Wewers
- Martin Woolley
- David & Jean Wrobleski