

# Pathways

## The Newsletter for Glycoprotein Storage Diseases

ISMRD, a 501 © not-for-profit organization, FEIN 53-2164838 | website [www.ismrd.org](http://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, Mucopolipidosis II alpha/beta (I-Cell Disease), Mucopolipidosis III alpha/beta (Pseudo-Hurler Polydystrophy), Mucopolipidosis 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



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### *ISMRD Board of Directors*

<b>President</b>	Jackie James
<b>Treasurer</b>	Mark Stark
<b>Vice President, Administration</b>	Jenny Noble
<b>Vice President, Research</b>	John Forman

### **Directors**

United States: Tish Adkins, Andrea Gates  
Australia: Carolyn Paisley-Dew, Juanita Van Dam

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## From the President's Desk

*By Jackie James  
President, ISMRD*



### **Greetings ISMRD family and friends!**

We have had a very busy and productive few months since our last newsletter. It is with much excitement that we introduce and give a very big welcome to our newest board member, Juanita Van Dam. In this newsletter you will hear a little of Juanita's story and background and her passion to raise funding for research for ML. As many of you know, ISMRD is striving to reach a minimum of \$100,000.00 toward ML research by June 2016. We are already half way there, but still have a way to go and we need your help to reach our goal. A huge thank you goes out to everyone of you that have donated funds. Your generosity and thoughtfulness is very much appreciated.

One of our amazingly talented ISMRD members, Katie Chika has created a whole line of ISMRD crafted items that you can purchase, and she is very generously donating the profits of those sales to ISMRD. Please check out our article on Katie and then check out her etsy store! We would like to thank Katie for all her hard work and generosity.

Rare Disease Day was held on February 29th of this year. Several ISMRD families participated in a tea where they either literally held a small tea and invited friends who donated funding, or where individuals donated the cost of what they would have spent on tea or coffee for that day. Others sold bracelets or had various events at schools and businesses. Thank you to each of you that participated! For your interest we have listed key facts and figures for events held around the world on this day.

Be sure to check out the information on what is happening around the world relating to conferences and research. It is very encouraging to see how scientists and researchers are focusing on our ultra rare diseases.

For our family that has recently lost a loved one to Fucosidosis, we would like to pass on our deepest and sincere condolences. Our thoughts and prayers are with you.

Please keep those that have had surgery as well as those that are facing surgery very soon in your thoughts and prayers. We have them listed toward the end of this newsletter

Keep your eyes on our website and Facebook page for updates and progress on the ML research initiative! Together we can make this happen!

Wishing you all the very best

**Jackie James**  
**ISMRD Board President**  
**Mom to Anna, ML III**



*Juanita Van Dam recently joined the ISMRD Board. She lives on the Gold Coast, Queensland, Australia and has two young children with Mucopolidosis. Here she tells her family's story.*

## Our Journey with Mucopolidosis by Juanita Van Dam

In December 2011, we attended a paediatric appointment to see if our son Damian had ADHD like his father and big brother. Our paediatrician looked at Damian and said 'I think he has something else'. Richard and I had no idea what he meant, we had no comprehension that those few words were about to change everything. He said 'Damian has coarse facial features, a big head and no nose bridge'. I looked at Damian trying to understand what he was saying. 'No! Damian has a gorgeous little man face and his big head is his Dutch heritage'. I hadn't even realised Damian had no nose bridge. How could we not see that? We literally had no idea that Damian and Jesse-Rose had such a serious degenerative rare disease.

I look at them both today, over four years later and they are gorgeous children with beautiful strong faces. Their personalities are amazing. Damian is the Joker. His smile is exactly the same as Jack Nicholson's in Batman. His laugh makes everyone around him laugh even when we have no idea what he is laughing at. He absolutely loves video games and will talk your ear off about them, including YouTube videos and how he is going to be a policeman when he grows up. He is exuberant and cheeky, lively and loud. He cannot walk very far without pain, but he will run himself ragged when there is fun to be had. He hates being different and will go to extreme lengths to not be different when at school, despite the pain he suffers afterwards.



Jesse-Rose is the light of our lives! She is funny, serious, cranky, moody, headstrong and very stubborn. Her smile lights up her face and makes everyone smile with her. She has this beautiful little dance that she has made up and she says to me 'Mum, I don't need dance lessons, I know how to dance!' She puts her gorgeous little hands on her hips and spins around. I watch with bittersweet sadness and joy, because she can't put her hands on her hips properly because they are stiff and chunky. She loves colouring in no matter how much it hurts her hands. She makes amazing drawings with cut out pictures. She tries to do absolutely everything herself, buttons, shoelaces, zippers, doing her hair. She also talks our ear off about everything, life, babies, what she is going to do when she grows up 'I am going to work with you Mummy!'

There is not a single day goes by that I don't get teary, thinking about this disease that is robbing them of their mobility and their quality of life. The fact that there is no cure and very little treatment. The fact that when they share what they want to do when they are grown up, it is highly unlikely it will happen. I see others going through this same journey and it is heartbreaking watching their pain and their parents'

helplessness at stopping their pain. We battle regularly with specialists to ensure our children get the checks that they need and we push for treatments that may have adverse side effects.

I look at them both sometimes and become absolutely gobsmacked at the seriousness of Mucopolysaccharidosis. Everything in my life has taught me to question authorities. Being a union delegate, political activist and pushing for change has led me to be exactly who I need to be for my gorgeous children. Since they have been diagnosed, Richard and I have had to question doctors, push for treatments and surgery, provide facts of the disease to specialists who have never heard of it before. We have become their advocates, medical activists and very pushy parents.

I have now joined the Board of ISMRD and felt very honoured to be asked. We have the opportunity to start research for Mucopolysaccharidosis, focussing on Type III. I have no idea if any research will be of benefit for Damian and Jesse-Rose, but it gives me hope. Hope to what the future holds. Hope that when they are adults they may be able to do what they talk to me about now. Hope that pain will not be the ruling factor in their quality of life and hope that they may have the chance of a cure.



### **ADDITIONAL BOARD MEMBER SOUGHT**

**ISMRD is seeking an additional Board member with Social Media Knowledge.**

**Help us get our message out to the world using Twitter, Instagram and Facebook.**

**Monitor these for us, and provide ongoing advice and assistance to help us lift the profile of ISMRD.**

**If this position is for you we look forward to hearing from you.**

**Please contact [info@ismrd.org](mailto:info@ismrd.org)**



## ISMRD's Mucopolidosis Research Initiative

***ISMRD's goal is to raise \$100,000 for Mucopolidosis research by June 2016. So far, we have raised \$80,000 which is made up of funds received, promises from other Partner organizations and other individuals.***

***Thank you for all your help with this initiative. We still need to raise a further \$20,000 by June 2016. Please help us by:***

- **Buying** from the ISMRD shop. Go to: <http://www.ismrd.org/fundraisers/ismrd-shop>
- **Fundraising.** For ideas, go to: <http://www.ismrd.org/fundraisers/fundraising-ideas>
- **Donating.** Go to <http://www.ismrd.org/>
- **Sharing** our request for donations with your Facebook friends.

<https://www.facebook.com/groups/82945687520/> (this is our closed to the public page)

<https://www.facebook.com/Mucopolidosis/> (this is open to the public)

**Please help us reach this very worthwhile goal! Every dollar helps!**



***Andre, Sergio, Zachie and Aiobhe  
all affected by Mucopolidosis***

## ISM RD MEMBER KATIE CHIKA MAKES AND SELLS HANDCRAFTED ITEMS, WITH ALL PROCEEDS TO ISMRD

Hi, my name is Katie Chika, my sister Emily has Alpha Mannosidosis. Emily is the youngest of 3 girls, Emily (21), Heather (26) and me (29). We all live in Illinois, USA. Emily was diagnosed with Alpha Mannosidosis at the age of 7.

Heather and I are both special education teachers. I am a self-contained teacher (special education teacher that teaches all academic subjects subjects) for students with learning and behavior disorders. Heather is a self-contained teacher for students with autism.



**Katie, Emily and Heather**

Emily loves going out with her sisters. Her favourite thing to do is go shopping at the Disney store and this past summer we went to Disney World, Florida.

In my spare time, I enjoy crafting and making personalized gifts for my family and friends. Many items have become very popular and I recently started an Etsy shop (Etsy is a peer-to-peer e-commerce website focused on handmade or vintage items and supplies, as well as unique factory-manufactured items - Editor). In my Etsy shop I have several different penguin items. I would like to donate profits back to ISMRD for any penguin item purchased. I can personalize items by adding names, changing colors, etc.

All of these items are hand made by me with permanent adhesive vinyl. With proper care, the vinyl will last anywhere from 7-10 years.

I have all sorts of ISMRD drink ware including travel cups, drink bottles, wine glasses (white, red, stemless), beer steins and Pilsner glasses. I can add a single penguin or a penguin family. I can put whatever you like on the back: ISMRD, ISMRD MOM, ISMRD DAD, ISMRD FRIEND, etc.



I also have car decals. All items can be customized and personalized.

**ALL PROFITS go back to ISMRD.**

There are 3 ways to order:

1. Find the link at <http://www.ismrd.org/fundraisers/ismrd-shop>
2. Directly from my etsy page at <http://www.etsy.com/shop/surfingintosped>
3. Message me at my Facebook page [www.facebook.com/katiescraftycorner2016](http://www.facebook.com/katiescraftycorner2016)

Please contact me with any questions! I am so happy to be able to help our ISMRD Family!

***Katie Chika***



Don't forget the ISMRD gofundme page, which has so far raised US\$4,501. 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 the ISMRD, at no cost to you. You'll find the same low prices, vast selection and convenient shopping experience as Amazon.com.

Go to <http://smile.amazon.com> for this easy way to help raise funds for the ISMRD.

**Shop at AmazonSmile**  
and Amazon will make  
a donation to:

Get started

**amazon**smile



## ISMIRD SHOP

We know time is precious for you all as you care for your special people and you often don't have time to do a fundraiser, but here is something really simple. Sell some of our on-line items to families, friends and work colleague's or why not direct them to our store [www.ismrd.org/fundraisers/ismrd-shop](http://www.ismrd.org/fundraisers/ismrd-shop)

**Every dollar counts toward our \$100K goal for ML Research.**



Advocate 4 ISMRD - bracelets \$2 each.



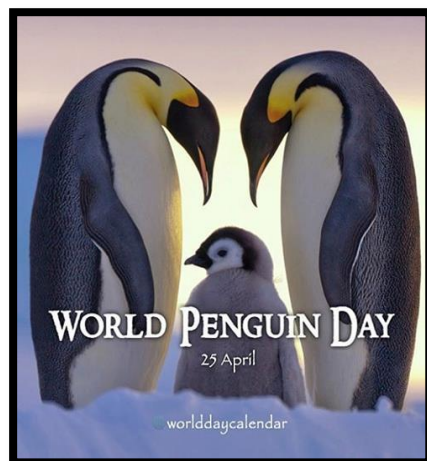
Penguin Bracelets  
\$20.00 each



ISMIRD Tee Shirts  
\$15.00 each



ISMIRD specially blended tea \$10.00 for 2 ounces







## Key facts and figures

### Events around the world

- Events were held in 85 countries and regions (all 28 EU countries!) with first time participants: [Andorra, Aruba, Indonesia, Libya, Mauritius, Moldova, Tanzania, Tunisia, Uganda and Zimbabwe](#)
- Hundreds of events organised by patient groups were displayed on Eurordis's [website](#)

### Participation and solidarity

- Policy makers were active in [Europe, Australia, Canada, Ireland](#) and [37 statehouse events in the US and more](#)
- Hundreds of organisations showed their solidarity by [becoming a Friend](#)
- Researchers, medical professionals, carers, healthcare employees and institutions [joined in to make the voice of rare diseases heard](#)

### The Rare Disease Day official video

- Published in 34 languages with over 300,000 views
- Disseminated worldwide via sharing on social media, particularly [Facebook](#)

### Rarediseaseday.org website

- A new look for Eurordis's website was launched
- 180,000 visits to [rarediseaseday.org](#)

### Photos, videos and testimonies

- The number of testimonies more than doubled on the website
- See pictures of solidarity around the world: ['Raise and Join Hands'](#)

### Media coverage

- Rare Disease Day was prevalent in print, radio, television and online all over the world
- Check out some of the things written around the world here on Eurordis's [media page](#).

### Social media

- 36,000 tweets with [#rarediseaseday](#) on February 29. The hashtag [trended](#) in the UK and US
- The [Thunderclap](#) campaign reached nearly 1.5 million people on social media
- Eurordis reached over 6 million people on [Facebook](#) alone and likes on Eurordis's page are now well over 77,000

Eurordis wants to hear about your events and experiences on Rare Disease Day. Upload your photos and videos onto their [website](#) and send them your press clippings and links to videos if you had any media coverage for their [media page](#)

If you haven't uploaded your 'Raise and Join Hands' photos you still have time. [Upload them here](#)

## Participants sought for Alpha-Mannosidosis Study

### *Reminder to all Alpha-Mannosidosis families*

The study being undertaken by Dr Stephen Kaler and his team at the National Institutes of Health in Bethesda, Maryland, has approval for an extension of the ages of eligibility. Patients with Alpha-Mannosidosis from age 5 to age 60 can now be included in this study which aims to establish biomarkers that can be used for later studies of gene therapy for this disease. Patients from ages 5 to 10 years of age are particularly welcome in the study.

Travel assistance and accommodation costs are provided by the NIH for study participants, and patients may be eligible whether transplanted or not. Please consider enrolling in this study to help accelerate the discovery of an effective treatment for Alpha-Mannosidosis.

Contact details are:

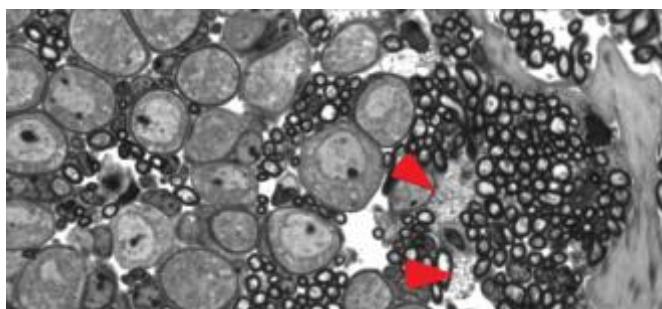
**Kristen Stevens, Research Nurse** [kristen.stevens@nih.gov](mailto:kristen.stevens@nih.gov) (301) 402-6103

**Stephen G Kaler, M.D.** [kalers@mail.nih.gov](mailto:kalers@mail.nih.gov) (301) 451-6034



## ISMRD is proud to be a sponsor of the Pathobiology of the Lysosome and Lysosomal Diseases Conference 2016

7th – 10th July 2016, at Clare College, Cambridge, UK



This conference will present an overview of recent progress in the understanding and management of lysosomal storage disorders. Internationally renowned scientists will cover subjects ranging from the basic biology and pathophysiology of this group of disorders to discussions of animal models, genetics, advancements in treatment, and pharmacoeconomic

challenges posed by rare diseases in general and lysosomal disorders in particular.

For more information, go to <http://www.zingconferences.com/conferences/the-metabolic-role-of-the-lysosome-and-nutrient-sensing/>



## What's happening around the world?

### Europe gives green light to first gene therapy for children

The world's first life-saving gene therapy for children has been recommended for approval in Europe, boosting the pioneering technology to fix faulty genes.

The therapy, called Strimvelis, developed by Italian scientists and GlaxoSmithKline, has been endorsed for a tiny number of children with ADA Severe Combined Immune Deficiency (ADA-SCID) for whom no matching bone marrow donor is available.

Around 15 children a year are born in Europe with the ultra-rare genetic disorder, which leaves them unable to make a type of white blood cell. They rarely survive beyond two years unless their immune function is restored with a suitable bone marrow transplant.

SCID is sometimes known as "bubble baby" disease, since children born with it have immune systems so weak they must live in germ-free environments.

Strimvelis is expected to secure formal marketing authorization from the European Commission in a couple of months, making it the second gene therapy to be approved in Europe, after UniQure's Glybera, which treats a rare adult blood disorder.

The U.S. Food and Drug Administration has yet to approve any gene therapies but a growing number of U.S. biotech companies have products in development.

Other large pharmaceutical companies are also eyeing the field.

**MANY SETBACKS:** Research into gene therapy goes back a quarter of a century but the field has experienced many setbacks, including the high-profile death of an American patient in 1999 and some disastrous clinical trial results in the late 1990s and early 2000s.

Now, though, optimism is building, helped by the discovery of better ways to carry replacement genes into cells.

Martin Andrews, head of GlaxoSmithKline's rare diseases unit, believes the technology is proving itself, although it remains at an early stage of development.

"We're on page one of chapter one of a new medicine text book," he told Reuters.

A host of challenges still need to be overcome, including the complexity of delivering a product like GSK's new treatment, which requires bone marrow cells to be taken from the patient, processed and injected back.

Trickiest of all may be pricing, given the tiny market for a therapy like Strimvelis. UniQure's Glybera made history in 2014 as the first drug to carry a \$1 million price tag. GSK is not putting a price on its product but a source close to the company said that, if approved, Strimvelis would cost "very significantly less than \$1 million".

GSK has several other gene therapies under development with researchers at Fondazione Telethon and Ospedale San Raffaele in Italy, including treatments for metachromatic leukodystrophy and Wiskott-Aldrich syndrome that could be submitted for regulatory approval in the next couple of years.

Its Strimvelis treatment for ADA-SCID is also being lined up for submission to U.S. regulators, although Andrews said this would not happen before the end of next year.

## European Conference on Rare Diseases 2016, Edinburgh



The 8th European Conference on Rare Diseases & Orphan Products (ECRD) will take place in Edinburgh from 26 – 28 May 2016.

ECRD 2016 will address current and future issues facing rare disease patients so that game-changing and sustainable responses can be developed.

With over 800 participants from more than 40 countries, ECRD 2016 provides a unique platform for all members of the rare disease community across all European countries: patients, patient representatives and caregivers; academics, scientists and researchers; payers and regulators; healthcare professionals, industry, policy makers and representatives of EU Member States.

Since the first ECRD 15 years ago, the conference has allowed the rare disease community to regularly gather to monitor relevant initiatives, drive the policy framework around diagnosis, treatment and care and to empower the community to drive change where it is needed most. Outputs from ECRD support continuity and extension of health and social policies on rare diseases across the EU, and at a national and local level.

## Rare Metabolic Disorders Conference, London

**Rare Metabolic Disorders:  
Detection, Research,  
Management and Treatment**

**20th - 22nd September 2016  
Location: London**

**EuroSciCon**

From congenital disorders to inherited metabolic diseases, this event will investigate recent developments in an informal academic setting, with an atmosphere conducive to debate and discussion. With a number of sessions over three days, many aspects of the metabolome will be covered, bringing together those working in academia, medicine, biotechnology and pharmaceuticals.

Presentation topics will include "Enzyme Replacement Therapy for Lysosomal Storage Disorders: the Pharmacology of Marginal Gains" by Dr Robin Lachmann of the National Hospital for Neurology and Neurosurgery in the United Kingdom.



## 4th Rare Diseases Summer School, Switzerland

**13-15 July 2016 Wädenswil (on lake Zurich), Switzerland**

The 4<sup>th</sup> Rare Diseases Summer School will focus on a wide variety of subjects in the arena of rare diseases, from disease mechanisms and animal models, to improving diagnoses, to novel therapeutics. There will be lectures and workshops on drug development, model organisms, how to choose clinical endpoints, clinical trials, regulatory aspects, patient registries, patient initiated research, ethical considerations, as well as what rare diseases may tell us about common diseases.

The summer school will contain lectures by national and international rare disease experts, workshops, and poster presentations by participants.

A high faculty-to-student ratio will allow participants to optimally benefit from the varied expertise of the faculty. There will be ample time for discussions, teamwork, as well as for socializing.

### Eligibility

The school addresses clinicians, physician-scientists, postdocs, and PhD students as well as select students from various disciplines such as medicine, biology, pharmaceutical science, psychology, sociology and related fields.



## First Book in Spanish on Ethics in Rare Diseases Research



A group of 14 authors from different disciplines have launched the book *Ethics in Research on Rare Diseases*, the first monograph on this subject published in Spanish, which was created with the aim of “responding to the ethical concerns that research in rare diseases currently poses”, according to the authors.

The work, signed by researchers, clinicians, regulatory agencies, bioethicists and family and patient representatives, addresses all ethical dilemmas involved in the investigation of these pathologies.

This book, sponsored by the Genzyme Foundation and the Center for Biomedical Network Research on Rare Diseases (CIBERER), addresses people directly or indirectly related to research in rare diseases, whatever their academic and professional training, both in Spain and Latin America.

[Ethics in Research on Rare Diseases](#)



## Carer Resources



### Genzyme Blog Series on Building Resilience for Rare Disease Caregivers

Vanessa King is an expert on positive psychology and resilience. As part of a blog series developed by Genzyme to help rare disease carers build resilience, she talks here about building your strengths, and the power of small steps

**Build Your Strengths by Vanessa King:** Being a patient group leader or carer means we draw on a wide range of skills and have to be good at many things. Yet how often do we take a moment to *notice* these capabilities, let alone think about how we can amplify them?

Research shows that when we identify our strengths and use them more and in new ways, we are happier. We have more energy We are more productive and more likely to achieve our goals - which enables us to be and feel more resilient.

**What are strengths?** We are all good at many things but not all of them are strengths. Our strengths are things that we naturally do well, learn easily, enjoy doing and, important, find energizing rather than depleting. They might be things that others see in us that we don't always notice in ourselves. This is because often these things are so natural for us we don't realize that they are something special, that not everyone can do.

There are many different strengths, for example: creative thinking, persistence, being organized, planning, thinking objectively, social skills, writing, story-telling, clear communication, kindness, attention to detail, making connections (between people, things or ideas), making things look nice, adaptability or seeing the bigger picture.

### **How do we identify our strengths?**

There are different ways we can identify our strengths:

#### 1. Reflect on your skills and capabilities:

- What come most naturally for you? What can't you help but use?
- What do you feel energized when doing? What would you say to complete the sentence: "I love doing....."?
- What gets done quickly?
- What do people know you and come to you for?

#### 2. Ask those around you (family members, colleagues, friends) to describe what they see as your top 3 – 5 strengths and when they see you using them. (Note: other people may see things that we are good at but we may not find these things energizing or enjoyable, so they may not be true strengths.)

#### 3. Take a strengths survey:

- Psychologists have developed several surveys we can take that help us identify our strengths. A good one to try is called the "Values In Action" survey (VIA for short). It's free to use. Click [here](#) to take it.

Aim to come up with your top 5, as it's easier to focus on a few rather than a lot of strengths. Make sure these are things that are genuinely "you" – things you most love to do and find energizing, not what you think you should be good at!

**Using our strengths more:** Like a muscle, we only get stronger if we intentionally practice using our strengths. So pick one of yours and take some time to think about how can you use this more and in new ways day-to-day. This doesn't necessarily mean doing whole new activities but thinking about how you do what you do. The more you focus on your strengths, over time, the more you will find new opportunities to use them. You can also try asking family members, colleagues and friends for their ideas on how you could apply your strengths in new ways.

The [attached guide](#) has some more tips and space to note down your thoughts on your strengths.

**Strengths in others:** In an earlier blog ([Keep Connected](#)) we looked at focusing on the strengths of others to help nurture our close relationships, so don't forget to practice spotting strengths in others too – the more you do it the easier it gets!

**Small Steps by Vanessa King:** As patient group leaders and carers, looking after yourself will help you look after others. It is very easy with all the demands on you to say “I just don't have time.” In this blog series we've explored lots of small and quick things we can do, which can make a big difference over time – for ourselves and for others.

**Taking control** Taking action, even the smallest of steps, has important psychological benefits. We've looked a number of small actions, for example [taking a moment to breathe](#), [focusing on the good things](#), or [moving more](#), that will benefit us directly.

Taking action gives us a sense of control, which is vital for our well being. There will be many areas in our lives where we don't have that much control – for example, the routines of the loved ones we are caring for, children's school times or rules at work, etc. Focusing on the areas that we do have control is important.

In this way, it doesn't so much matter which action you decide to take to boost your own resilience and well being, simply the act of doing something regularly will have a positive benefit. It will boost our self-confidence in our ability to make a difference for ourselves (what psychologists call “self-efficacy”). This then tends to have a positive ripple effect to other areas of our own lives and that of others.

**Small steps:** Recently psychologists discovered that one thing that makes a big difference to whether we have a good day or a bad day is feeling a sense of progress. This doesn't just mean achieving big goals we may have set ourselves, but tiny micro steps on a day-by-day basis. For example, many of us will recognize the good feeling we get from ticking things off our to-do lists.

So, as a carer or patient group leader or both, even if you don't have much time or head space, think what could you do in 10 minutes, 5 minutes or even 1 minute that will help you maintain your wellbeing and build your resilience. After all, we don't get to the top of the mountain without taking small steps.

The [attached guide](#) gives some more ideas on taking action and a planning sheet to help (writing goals down make it more likely you will do them!).

## Educational Toolbox on Medicines Research and Development in seven languages

The [European Patients' Academy \(EUPATI\)](#) has announced the launch of its new online [Toolbox on Medicines Research & Development](#). Designed for patients, patient groups, and anyone interested in learning more about medicines research and development (R&D), the freely-accessible toolbox includes materials covering drug discovery, clinical development, regulatory affairs, medicines safety, pharmacovigilance, and health technology assessment. The toolbox is available in seven languages: English,



French, German, Spanish, Italian, Polish and Russian. It was designed by patients in collaboration with experts from other stakeholder groups such as academia and industry. Users can access a wealth of fact sheets, graphics, slideshows, videos, recorded webinars, print-ready materials as well as a full glossary.

*"The toolbox is a comprehensive, self-explanatory, educational resource that has been built so that learnings on medicines R&D can be developed and shared by patient advocates. It is the result of a long-term, concerted effort by expert stakeholders including patients, researchers and academics. Our vision is that thousands of patient advocates in Europe will leverage the toolbox to enable more meaningful patient involvement and partnership in medicines R&D."* says Jan Geissler, Director, European Patients' Academy.

The need for the new toolbox is backed up by [recent EUPATI research](#) which reveals that pharmaceutical industry personnel believe patients can improve medicines research and development.



## Southwest Airlines offers free medical flights to families

Southwest Airlines has announced that it will offer \$3.2 million in free flights to families in need of specialized care at 101 facilities around the country.

Rady Children's Hospital and Scripps Health are among the nonprofit healthcare institutions taking part in Southwest's 2015 medical transportation grant program.

"We care about the communities we serve and are honored to partner with like-minded hospitals and medical transportation organizations," said Linda Rutherford, Southwest Airlines' vice president of communication and outreach.

"We are proud to help fill the gap between where patients live and where the doctors they need practice so patients can receive the treatment that best supports them," she said.

Airline officials said it will be easier for patients to get high-quality healthcare when they don't have to worry about transportation costs. The \$3.2 million total is 14 percent higher than last year.

Families interested in participating in the 8-year-old program are asked to contact their hospital's social work, travel/concierge or patient assistance offices, according to Southwest.



## Free Summer Camp for Families in Northeast USA 2-5 June 2016

NORD, in partnership with The Hole in the Wall Gang Camp, established by the late actor Paul Newman, is offering its first-ever rare disease camp, to be held in Connecticut.

This special Summer Family Camp is for children and families impacted by rare diseases to join together for a weekend of pure fun – free of charge. The Hole in the Wall Gang Camp will be hosting the camp for 25 families in the Northeast Region of the US.




Families who are located in the Northeast Region of the US. Are encouraged to apply! The application includes a portion for general information, family medical and consent forms for each member of the family who is attending, and a medical exam form to be completed by the diagnosed child's medical provider.

Apply now to attend this weekend of pure fun – free of charge. The camp is limited to 25 families.



ISM RD WARMLY WELCOMES YOU  
TO OUR FAMILY



-  Chris Knaus Black
-  Adalynn Cole and her family. They live in Michigan USA. Adalynn is four years old and was recently diagnosed with Fucosidosis.
-  Hermance Incardona and her family. They live in France, and Hermance has Aspartylglucosaminuria.

With Deepest Sympathy



May the stars carry your sadness away,  
May the flowers fill your heart with beauty,  
May hope forever wipe away your tears

**We mourn the loss of**

**Mercedes Arreguin who passed away on 1 March 2016 aged 12 years.**

**Mercedes had Fucosidosis, and lived in New Mexico USA.**

**Our deepest sympathy goes out to her family.**



## An Angel's Hand



An angel whispered in my ear  
Believe in me and have no fear,  
Come with me and take my hand,  
I will take you to another land.  
A place of peace, a place of light,  
Don't be afraid, just hold on tight.  
Your Earth days are done but don't be sad  
Remember all the times you had;  
Loved ones will join you, don't despair.  
You have many in Heaven waiting there.  
Your body dies, your soul is free,  
That's why you get to fly with me.

By John F. Connor





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](mailto: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**

- 🌱 Sam Jamil, who lives in Manchester in the UK and had dental surgery
- 🌱 Skylar Thomas, who lives in Texas USA and had a total knee replacement, Skylar has Mucopolysaccharidosis III
- 🌱 Savahna Amour who lives in Arizona USA and has Chronic Obstructive Pulmonary Disease
- 🌱 Kristopher Gaffner, who lives in Michigan USA and had surgery for pleural effusion. Christopher has Alpha-Mannosidosis
- 🌱 Sarah Noble who lives in New Zealand is having Spinal surgery and has ML III alpha/beta

## ISMARD'S Sunshine Care Committee



**ISMARD 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**

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