

# 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 and 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

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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



### ISMRD Board of Directors

<b>President:</b>	Mark Stark
<b>Vice President, Administration:</b>	Jenny Noble
<b>Vice President, Fundraising:</b>	Pam Tobey
<b>Vice President, Research:</b>	John Forman

### Directors

United States: Jackie James | Andrea Gates | Susan Kester  
Tish Adkins

Australia: Carolyn Paisley-Dew

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## From the President's Desk

*By Mark Stark  
President, ISMRD*



Greetings to you and your family. Wherever you are in the world, I hope you are experiencing good weather and good health.

I would like to extend a warm welcome to the 15 new families that have recently joined the ISMRD. They represent six countries, three continents and seven of the nine Glycoprotein Storage Disorders. Please go to page 19 of the newsletter to see a list of our new members.

Early bird registrations for our conference have now closed, but we are more than willing to consider this for families who still wish to attend but have not yet registered. If you are still considering attending and would like the option of early bird registration please contact us at [info@ismrd.org](mailto:info@ismrd.org) to discuss.

We are very keen to complement our Board with two additional members who can assist with communications and fundraising. Please see page 4 for details. We would welcome any questions you may have.

Three of our Board members attended the 11th Annual World Symposium, and in this newsletter there are some very interesting posters that were presented at the Symposium.

One of our Board members achieved a coup with the Commonwealth of Virginia (see page 14).

Also presented in this edition is an interesting array of developments in Europe and Australia for people with a rare disease, and also some resources for our hard-working carers.

I hope you will enjoy this issue of the ISMRD newsletter.

Thank you  
**Mark Stark**  
**President**  
**ISMRD**



## ISMRD is seeking new Board Members

We are seeking two people to step into the following roles:

**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.

Please note that these and 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](mailto:info@ismrd.org)



ISMRD Board Members John Forman, Jenny Noble and Susan Kester attended the WORLD Symposium in Florida in 2015. World Symposiums are held annually and this year's Symposium was designed to provide an interdisciplinary forum to explore and discuss specific areas of interest, research and clinical applicability related to lysosomal diseases. The Symposium was designed to help researchers and clinicians to better manage and understand diagnostic options for patients with storage diseases, identify areas requiring additional basic and clinical research.

Here are the following posters from the conference:

1. Pharmacological chaperoning in Fabry and Schindler diseases;
2. Is Melanogenesis disturbed in Mucopolidosis II/III? A multicenter study based on clinical and genetic findings;
3. Long-term neuropsychological follow-up in a patient with  $\alpha$ -Mannosidase;
4. Molecular basis of Sialidosis and its treatment;
5. Updates in biochemical and molecular diagnosis of Brazilian patients with Mucopolidosis II/III alpha/beta;
6. Updates in biochemical and molecular diagnosis of Brazilian patients with Mucopolidosis II/III alpha/beta.

***Below are a number of Poster abstracts that caught the attention of the Board.***

## **Pharmacological chaperoning in Fabry and Schindler diseases**

*Scott C. Garman, Matthew C. Metcalf, University of Massachusetts Amherst, Amherst, MA, USA*

Two homologous lysosomal enzymes include  $\alpha$ -galactosidase ( $\alpha$ -GAL) and  $\alpha$ -N-acetylgalactosaminidase ( $\alpha$ -NAGAL), which are deficient in the lysosomal diseases known as Fabry and Schindler diseases, respectively. One approach for the treatment of the diseases uses pharmacological chaperones, or small molecules that bind to the enzymes and confer stability to the folded form of the proteins. We have determined the three-dimensional structures of the  $\alpha$ -GAL and  $\alpha$ -NAGAL enzymes, both alone and in complex with a range of pharmacological chaperones. In this work, we will present the molecular basis for pharmacological chaperoning in the family of enzymes that include  $\alpha$ -GAL and  $\alpha$ -NAGAL. Using rational design approaches, we were able to engineer affinity improvements of more than 1 million fold for small molecules that bind and stabilize the enzymes. We will present guidelines for improving the specificity and affinity of compounds for these and other lysosomal enzymes.

## Is Melanogenesis disturbed in Mucopolidosis II/III? A multicenter study based on clinical and genetic findings

Mucopolidosis (ML) II and III are inborn errors of metabolism caused by deficient activity of GlcNAc-1-phosphotransferase, an enzyme responsible for targeting of lysosomal hydrolases to the lysosomes. We hypothesized that melanogenesis would be altered in patients with ML II and III.

**Objectives:** **1)** To characterize the skin, hair, and eye color of patients with ML II and III and compare these features to healthy controls; and **2)** to establish a genotype–phenotype association involving SNPs known to be associated with skin, hair, and eye color in normal populations.

**Methods:** This multicentre, prospective, controlled, cross-sectional study employed a convenience sampling strategy. Brazilian patients with ML II and III aged  $\geq 1$  year and their parents, as well as healthy controls based in samples of the CANDELA project, were examined for skin, hair, and eye characteristics using standard (such as the Fitzpatrick scale) and/ or visual classifications. Patients were screened for SNPs [rs1126809 (TYR gene), rs16891982 (SLC45A2 gene), rs1426654 (SLC24A5 gene) and rs1129038 (HERC2 gene)] through KASP genotyping assay.

**Results/discussion:** Seventeen patients (ML II= 7, ML III alpha/ beta=7, ML III gamma=3; 13 male, 4 female; mean age,  $13 \pm 12.5$  years) and 29 parents were included in the study, as well as 185 healthy subjects from CANDELA project. Most patients had Fitzpatrick skin types I–III ( $n = 14/17$ , 82%) a rate discrepant with the skin type of their parents ( $n = 19/29$ , 66%). One of our patients (Fig. 1) had Fitzpatrick skin type II (blond hair, blue eyes, fair skin) at the age of 2 years, while both parents were skin type IV, corroborating the biological plausibility of skin pigmentation changes as manifestations of ML II and III (Figure). Regarding genotype–phenotype association, for rs1126809 2/17 had lighter hair than expected; for rs16891982, 4/17, 6/17 and 10/17 patients had, respectively, lighter eyes, hair and skin color, and 3/17, 4/17 and 2/17 patients had, respectively, darker eyes, hair, and skin color than expected; and for rs1426654, 2/17, 1/17, and 1/17 had lighter eyes, hair and skin and 9/17 and 3/17 had darker hair and skin than expected; and for rs1129038 6/ 16 patients had darker eyes than expected. Any significant discrepancy was not found regarding the eye and hair color predicted by genotype and those one found in healthy controls.

**Conclusion:** Patients with ML II/III appear to exhibit changes in melanogenesis (most commonly hypomelanosis). Further studies are required to corroborate these findings.

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## Long-term neuropsychological follow-up in a patient with $\alpha$ -Mannosidase

*Johanneke B. Ebbink, Femke K. Aarsen, Esmee Oussoren, Iris Plug, Hannerieke J.M.P. van den Hout, Ans A.T. van der Ploeg, ErasmusMedical Center, Rotterdam, Netherlands*

$\alpha$ -Mannosidosis is a very rare lysosomal disorder resulting from deficiency of  $\alpha$ -mannosidase. No therapy was available until now; a phase III study on the effect of enzyme replacement therapy is currently underway. The limited studies on the natural course of cognition show intelligence in the range from mild mental development to profound mental retardation. Studies on long-term intellectual development

present inconsistent data and there are no studies on neuropsychological development. With this case report, we present a 16 year neuropsychological follow-up of an untreated Caucasian patient. The patient had a mild developmental delay which remained stable in time (range IQ 71–81; 8 measurements between age 1 and 17 years). At last assessment his mental age was 8.0 years (chronologic age 17.9 years). Three additional neuropsychological tests showed difficulties in attention, memory and fine motor skills (age 9, 14 and 17 years; data was corrected for intelligence). His fine motor skills and memory

deteriorated in time. School performances were in accordance to his intelligence. Behavioral questionnaires filled out by the parents and an interview according to DSM IV criteria showed symptoms of pervasive development disorder (PDD, not otherwise specified). Brain MRI at age 16 showed mild atrophy in the cerebrum and cerebellum. He had diminished myelination of the periventricular white matter. Here we report a patient with alpha-mannosidosis having a subnormal intelligence, which remained stable in time. The patient performed relatively well on his intelligence tests, when compared to literature on cognition. So far, our case report adds to the existing literature that alphasialidosis is not a progressive mental disorder. The patient had difficulties in his attention, finemotor skills and memory. His finemotor skills and memory deteriorated in time. The impairment in memory and attention was not reported before. With the phase III trial underway, solid knowledge of the natural course and the specific neuropsychological deficits of the disease is important, especially of mental development which is the main characteristic of the disease.

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## Molecular basis of Sialidosis and its treatment

*Nilima Kolli, Scott C. Garman, University of Massachusetts Amherst,*

*Amherst, MA, USA*

Sialidosis is a lysosomal disease caused by mutations in the lysosomal neuraminidase (NEU1). NEU1 is an exosialidase that cleaves terminal sialic acids from glycoproteins and glycolipids in the lysosome and on the cell surface, highlighting its importance in vital cellular functions such as catabolism and signal transduction pathways. Deficiency of NEU1 activity in patients with mutations in Protective Protein/Cathepsin A (PPCA, galactosialidosis) suggests its dependence on PPCA for normal function. In the current study we show that PPCA directly interacts with NEU1 and, contrary to previous reports, we demonstrate that this interaction is not required for the catalytic activity of NEU1, instead it facilitates stabilization of NEU1. We report for the first time the kinetic parameters of the neuraminidase activity of NEU1, which exhibits a  $k_{cat}$  of 2.14  $\text{min}^{-1}$  and  $k_{cat}/K_M$  of 1.73  $\text{mM}^{-1} \text{min}^{-1}$ . Using inhibitors to stabilize NEU1, we further present a proof-of-concept study for pharmacological chaperone therapy for the treatment of sialidosis. Thus, we demonstrate that instability of NEU1 outside lysosomal complex forms the molecular basis of sialidosis, and propose development of therapeutics focused on stabilization of NEU1.



## Updates in biochemical and molecular diagnosis of Brazilian patients with Mucopolipidosis II/III Alpha/Beta

Fernanda S. Ludwig, Taciane Alegria, Nataniel Ludwig, Maira G. Burin, Ida V.D. Schwartz, HCPA/UFRGS, Porto Alegre, Brazil

Mucopolipidosis II and III (ML II and III) alpha/beta are autosomal recessive lysosomal disorders (LSD) in which the essential mannose 6- phosphate recognition marker system is deficient. They are described as being caused by mutations in GNPTAB gene, which encodes alpha/beta subunits of N-acetylglucosamine-1-phosphotransferase (phosphotransferase). The biochemical diagnosis is usually based on indirect biochemical method: measurement of several lysosomal hydrolases activities. However, there is no consensus about which enzymes should be investigated, and different panels are used with weak evidence to support each option. The molecular diagnosis is based on sequencing all exons of the gene, which contains 21 exons.

**Objective:** Report of advances in biochemical and molecular diagnosis in Brazilian patients with ML II/III alpha/beta through the proposal of a more rational panel for lysosomal hydrolases analysis and a protocol for identification of GNPTAB mutations to be applied in the hierarchy of exons.

**Methods:** The analysis was a retrospective study including patients with a clinical and biochemical or genetic diagnosis established at the reference center for diagnosis of lysosomal disorders in Southern Brazil. We developed a score based on 11 items to choose which enzymes should be included in the panel. To establish a molecular protocol, we analyzed mutations found in Brazilian patients with ML II/III alpha/beta investigated by our group through sequencing of GNPTAB with genotype identified.

**Results:** A total of 26 patients were included (MLII=14, MLIII=3) in biochemical analysis. In plasma, all enzymes exhibited high mean activity, except for chitotriosidase; in fibroblasts, only mean  $\beta$ -glucosidase activity was normal, with all others reduced. Comparing ML II with ML III,  $\alpha$ -N-acetylglucosaminidase level in plasma was higher and  $\beta$ -glucosidase level in fibroblasts was lower in ML II. The highest scores (e.g., better scores) were assigned to  $\alpha$ -mannosidase (29  $\alpha$ -l-iduronidase (28 points), total  $\beta$ -hexosaminidase (27 points),  $\beta$ -glucuronidase (26 points), and  $\alpha$ -N-acetylglucosaminidase (25 points). In molecular diagnosis, 16 patients were analyzed. Twelve different mutations were found in patients analyzed, being the most frequent the c.3503\_3504delTC (allele frequency=37.5%, exon 19). The exons with a higher rate of pathogenic variations found were exon 19, exon 13 (20% of alleles), and exon 10 (12.5% of alleles), followed by exons 3, 12, and 14.

**Conclusion:** For the biochemical analysis, we suggest the measurement of plasma and fibroblast levels of at least three of the enzymes which had the highest scores in our analysis. For the molecular diagnosis, we suggest that exon 19 of GNPTAB should be the first one to be analyzed, followed by exons 13 and 10, and, finally, by exons 3, 12, and 14. If we do not find any mutation in these exons, the remaining exons should be analyzed. The proposed panel of enzymes would be a faster, simpler, and less expensive method for the biochemical diagnosis of ML II and III, and the determination of a protocol analysis for DNA diagnosis has great importance, since it decreases the time employed in the analysis and allows a significant reduction of costs assigned.

## Alpha-Mannosidosis and compassionate use of Alpha-Mannosidase

### (Lamazym™): Two case reports

*Monica Lopez-Rodrigueza, Jesus Lacasa-Marzoa, Gema Perez Martina, Valerio Delgado-Cirerola, Enrique Berrocal-Valenciaa, Olga Tornero-Torresa, Raquel Fuentes-Irigoyena, Mercedes Gil Camposb, aHospital Central Cruz Roja, Madrid, Spain, bHospital Universitario Reina Sofia,*

*Cordoba, Spain*

Alpha-mannosidosis is a type of oligosaccharidosis due to a deficiency in the activity of acid alpha-mannosidase enzyme. The implicated gene is MAN2B1. This condition is inherited in an autosomal recessive pattern. And it is estimated to occur in approximately 1 in 500,000 people worldwide. The main clinical features are: Skeletal deformities, with dysostosis multiplex and coarse facies; intellectual disability, with behavioral changes, neurological symptoms including difficulty in coordinating movements (ataxia); and immune deficiency (frequent and severe bacterial infections). We report the cases of two siblings (non-consanguineous parents), diagnosed of alpha-mannosidosis in their early childhood, with an attenuated form (type II). The first case is a male, 22-year-old, with moderate intellectual disability, hip dysplasia, ataxia, neurosensorial deafness and coarse facies, and severe sepsis in his childhood (Fig. 1). The other case is a female, 18 year-old, with moderate–severe intellectual disability with behavioral problems, ataxia, myopia, neurosensorial deafness and dysostosis multiplex with coarse facies (Fig. 2). In April 2013, they were enrolled in a phase III clinical trial: rhLAMAN-05 (A multi-center, double-blind, randomized, placebo controlled, parallel group trial, investigating the efficacy and safety of repeated Lamazym treatment of subjects with alpha-mannosidosis), during 52 weeks, receiving the enzyme weekly by endovenous infusions. When the clinical trial was completed in April 2014, and given no safety concerns have arisen, the patients were offered to receive Lamazym™ as post-study therapy in their own hospital, as a compassionate use. During this period, the weekly infusions have been very well tolerated, with no reactions. Nowadays, it is still very soon for knowing the effectiveness of this enzyme replacement therapy. The outcomes of the trial were: Reduction of oligosaccharides in serum, the number of steps climbed in 3 min (3-minute stair climb test), forced vital capacity, the distance walked in 6 min (6-minute walk test). But we are able to confirm the excellent drug tolerance and the no appearance of severe adverse reactions during the follow-up period. It is necessary to know the trial results, to assess the efficacy and its posterior use in our clinical practice.







## ISMRD Fundraisers

### Kylie and Kendall Sweet 16 Celebration



### A message from Jamie, Kelly, Colin and Cayden

Our beautiful girls Kylie and Kendall passed away at age 8 and 5 from Mucopolysaccharidosis II, or I-Cell Disease. Since the girls' diagnosis, ISMRD has become a huge factor in helping families with I-Cell children by supporting them, connecting us with other families, and also funding research with hope that some day there will be a cure for this terrible disease. Please join us at our event supporting ISMRD and helping those families that continue to travel this journey.

**Kylie and Kendall Sweet 16 Celebration**  
**Saturday June 27th, 5:00 pm-closing**  
**The Golf Farm, aka Deacons Bar and Grill, 2100 US-12, Wauconda, IL**

Donations can also be made directly through [this site](#) or mailed to 2817 N. Sterling Drive, McHenry, IL 60050 c/o KandK Fundraiser.

Thank you for all of your support!!

## Raise money for your conference accommodation costs

### Beautiful bracelet in several colours



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](mailto: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.



The ISMRD gofundme page has now raised \$4,326. If you would like to donate, go to <http://www.gofundme.com/5rpjhw> Also please share this link with your network and say a few words to encourage them to donate. Every little bit helps and is gratefully received.



### 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 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

amazon smile

**SALE**

**ISMRD 2015 Calendars**

**USD\$9**



Reduced from USD\$14, the calendar features more of our kids than last year. Includes postage.

See a calendar [here](#)

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.



**To all our ISMRD mothers  
in countries celebrating Mother's Day on Sunday 10 May 2015**

Did you know that Mother's Day was started by American woman Anna Jarvis in 1908, and that she tried to get it closed down when it became commercial? Some countries use a different day of the year to coincide with an existing day of celebration e.g. in the United Kingdom, Mother's Day was incorporated into Mothering Sunday, celebrated on the fourth Sunday of Lent in March or April. For a more detailed history of Mother's Day, and to see when other countries celebrate it, go to

[http://en.wikipedia.org/wiki/Mother%27s\\_Day](http://en.wikipedia.org/wiki/Mother%27s_Day)





## What's happening around the world?

### Rare Disease Day - Virtual Morning Tea

ISMRD held a Virtual Morning Tea to celebrate International Rare Disease Day, 28 February 2015. Members from several countries participated. Some held actual morning teas and others donated the cost of their staff morning tea or the cost of something they purchased. At ISMRD Board member Susan Kester's workplace, all the staff came in Jeans and donated a gold coin. Over \$400 was raised from the Virtual Morning Tea. We hope that next year even more members will take part.



ISMRD Board member Jenny Noble held a morning tea in Tauranga, New Zealand for Rare Disease Day. A wonderful time was had by all with lots of conversation and yummy food.

## Rare Disease Day for the Commonwealth of Virginia




ISMRD Board member Tish Adkins approached Virginia Governor Terry McAuliffe (D), who has now declared February 28, 2015 as Rare Disease Day for the Commonwealth of Virginia.

Well done Tish!



## Austrian National Action Plan for Rare Diseases


 Austria has recently adopted its National Action Plan for Rare Diseases, is called NAP.se. The NAP.se contains nine key topics:

- Documentation of rare diseases in the health and social system;
- Improvement of medical-clinical care of rare disease sufferers;
- Improving the diagnosis of rare diseases;
- Improving the treatment of and access to treatments for rare diseases;
- Research in the field of rare diseases;
- Improving knowledge and awareness of rare diseases;
- Improving epidemiological knowledge in the context of rare diseases;
- Establishment of permanent consultative bodies for rare diseases; and
- Recognition of the benefits of self-help.

**Read more:** [Austrian National Action Plan for Rare Diseases](#)



## Czech Action Plan for Rare Diseases

 A ten-year Czech National Strategy for Rare Diseases was approved by the Czech government in 2010 for the country's rare disease patients, estimated to be roughly 20,000 out of 10.5 million habitants.

In the National Action Plan for Rare Diseases for the period 2015-2017 (NAP2) emphasis is placed on the support of early identification and diagnosis, on the further centralisation of care for rare disease patients, the unification and development of data collection and the creation of standards of care for patients with rare diseases, as well as on the improvement of awareness and education in rare diseases on continuity with the already established foreign cooperation and on establishing new contacts, and on the connecting Czech rare disease centres with common European databases and registers aggregating clinical and laboratory data. An emphasis is also placed towards joining international rare disease research projects domestic research projects as part of the Agency for Healthcare Research and the development of cross-border diagnostic and medical care in accordance with the provisions of the Directive on Patients' Rights in Cross-Border Care, which was transposed into Czech legislation in 2014.

The strategy also includes plans for the establishment of centres for home care, social subsidised hospital beds, respite centres and rehabilitation facilities, and the expansion of long-term care beds with trained personnel and special equipment to improve the quality of life and social inclusion of rare disease patients. The strategy also asks for attention to be placed toward effective and timely pharmacotherapy of rare disease following internationally-accepted standards and recommended approaches, and increasing the education and awareness on RD issues among the medical professionals and the public at large.

**Read more:** [The Second Czech National Action](#)

## New Paediatric Institute for Rare Diseases in Barcelona, Spain



Sant Joan de Déu-Barcelona Children's Hospital, in Barcelona, Spain has launched a Paediatric Institute for Rare Diseases (IPER) which will offer unified medical care for patients with rare diseases. The aim of IPER is also to obtain a quick diagnosis for the children who do not currently have a diagnosis. IPER will concentrate its efforts on translational research into diagnosis and treatment of rare diseases as soon as possible.

Read more: [CIBERER website](#) (in Spanish)



## Ireland's first National Rare Disease Office



Ireland's first National Rare Disease Office is expected to open officially in the second quarter of 2015. The establishment of a National Rare Disease Office (NRDO) was one of the key recommendations of the Department of Health's first national rare disease plan, '[A Rare Disease Plan for Ireland 2014-2018](#)', which was published last July.

The primary benefit of the new National Rare Disease Office will be the streamlining of access to diagnosis and treatments for patients. The new office will be responsible for building information on the availability of expertise in Ireland for rare diseases and making this information available to both patients and doctors. It will also establish a helpline for patients with access to a genetic counsellor, support the HSE in the mapping and validation of centres of expertise in rare diseases in Ireland, as well as having a long-term role in rare diseases surveillance.

Read more: [Irish Rare Disease Plan](#)

[Irish National Rare Disease Office](#)



## Australian Rare Diseases Summit



ISMRD Board Members John Forman (New Zealand) and Carolyn Paisley-Dew (Australia) Attended the Australian Rare Diseases Summit in Melbourne, Australia in March this year. The Summit represents a huge step towards a National Rare Disease Plan for Australia.



# Carer Resources

## Athena Diagnostics program for more affordable diagnostic testing for US patients

Athena Diagnostics has recently announced a new program to make diagnostic testing more accessible and more affordable. On September 1, 2014, the company launched the Athena Alliance Program to expand patient access to a variety of diagnostic methodologies and tests, especially those for rare and esoteric disorders.

Athena's priority is providing patient-centric customer service so that each patient has an individual specialist and a team of dedicated personnel to support them from the time of the order through test results. Athena Diagnostics offers a comprehensive test menu and intellectual property portfolio for neurological, neuromuscular, endocrine, and renal conditions through more than 400 diagnostic tests.

For individuals and their providers who have faced financial barriers to accessing the diagnostic tests necessary to make diagnostic and treatment decisions, the Athena Alliance Program may offer what you need.

If the physician has determined that it is medically necessary for the patient or a family member to receive a laboratory test for a rare or neurological disorder, have them contact the Athena Alliance Billing Team to engage one of their specialists at (800) 394-4493.

For more information, go to <http://www.AthenaDiagnostics.com/alliance>.



## Rare Diseases: A Compassionate Guide For Teenagers For End-Of-Life Care

Aging with Dignity, a Tallahassee, Florida-based non-profit organization published in 2012, a guide titled, "[Voicing My Choices: A Planning Guide for Adolescents & Young Adults](#)", that helps adolescents and young adults identify the choices that they can decide upon when faced with a rare disease or terminal illness. The guide is designed to aid them with communicating their preferences and desires to friends, family, and caregivers. It helps to express:

*"your thoughts about how you want to be comforted, supported, treated, and remembered ... it was developed using feedback from young people living with a serious illness ...".*

In September 2014, the Institute of Medicine published a Report, "[Dying in America](#)", that shows that adolescents are competent enough to talk about their end-of-life wants and that they prefer to be engaged in conversations about their care.

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## Blog Series on Building Resilience for Rare Disease Caregivers

In 2014, Genzyme supported the development of an [online resource](#) tailored to rare disease caregivers. They also they hosted two Caregiver Resilience Building workshops with patient group leaders in Europe facilitated by Vanessa King, an expert on positive psychology and resilience. These workshops were so successful that Vanessa will be a guest blogger over the course of 2015. She will share a number of different ideas and activities which have all been scientifically shown to boost wellbeing and resilience.

Go to [Caregiver Action Network](#)

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## A GPS for Families of and Individuals with Disabilities

New Jersey's Statewide Parent Advocacy Network Inc (SPAN) has developed a very comprehensive and useful guide for families of people with special needs and the professionals who work with them.

Go to: [A GPS for Families of and Individuals with Disabilities](#)



## Accessible Denim

**ABL Denim** make jeans for people who use a wheelchair as well as for kids who have sensory issues.

Their jeans are made with a higher back to accommodate sitting in a wheelchair and they feature a pocket on the lower leg to make storing a phone or wallet more accessible.

The kids' versions can be ordered without any metal (zippers or rivets) and with reversed seams around the waist so as to not cause irritation. The company is based in the US and will ship overseas. N.B. shipping costs are quite high: Canada \$25; Western Europe \$50; Australia \$45. Read more: [ABL Denim](#)



## ISMRD warmly welcomes to our family



- 🌸 Greg, Nancy and Jeremy Gatta. Jeremy has Mucopolysaccharidosis III. They live in Michigan, USA
- 🌸 Shirley Jamil and son Sam, who is 12 years old and has Mucopolysaccharidosis II. They live in the UK
- 🌸 Ana María Palma Ahumada and Felipe Crisostomo Ortiz, who live in Chile, South America
- 🌸 Deni Martin, who lives in Rio Negro, Argentina
- 🌸 Mike Schleiter who lives in Michigan, USA
- 🌸 Mary Blomberg and daughter Karin, who has Aspartylglucosaminuria. They live in Washington, USA
- 🌸 Maggie Hunt, who lives in Longford, Ireland
- 🌸 Silvia Blanco and her 18 month old son who has Mucopolysaccharidosis II
- 🌸 Alisha and Juan Arreguin whose child Mercedes has Fucosidosis. They live in New Mexico, USA
- 🌸 Andrea Schwartz and her daughter Helena who is four years old. Helena has Fucosidosis. They live in Austria
- 🌸 Naty Martino who lives in Argentina and has Mucopolysaccharidosis III
- 🌸 Laurel Mills whose child Oliver has Beta Mannosidosis and lives in the USA
- 🌸 Susan Faucher whose daughter Darla Marie has Alpha Mannosidosis. They live in the USA
- 🌸 Lucas Todres whose daughter has Sialidosis. They live in Argentina














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 ill or in hospital**

**Please know that the Penguin family thoughts and prayers are with you**

-  Bianca Trestianu, Mucopolysaccharidosis II
-  Mary Sibert, Mucopolysaccharidosis III
-  Alli Dennis, Mucopolysaccharidosis III
-  Skylar Thomas, Mucopolysaccharidosis III
-  Dale Lee, Alpha Mannosidosis
-  Heather Scott, Mucopolysaccharidosis III
-  Faith Patriquin,



# 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.



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

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