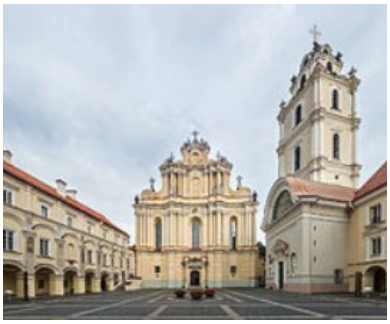
FABRY
International Network

Newsletter

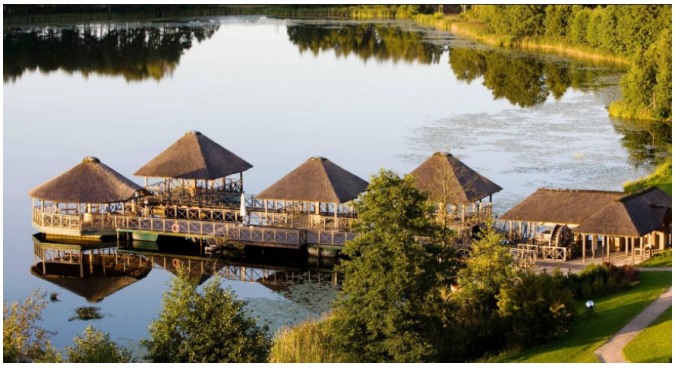
December 2017

Contents

FIN Fabry Expert Meeting
Vilnius, Lithuania
8th & 9th June 2018



The Board of the Fabry International Network are pleased to announce the next Fabry Expert Meeting will be held in Vilnius, Lithuania.



Confirmed speakers:

Professor Atul Mehta

Dr Rick Steeds

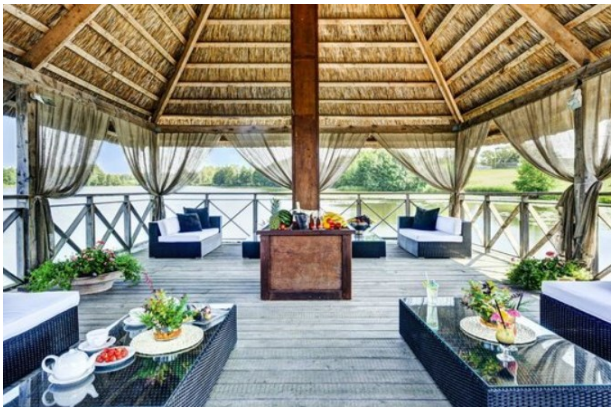
Dr Saunder-Plassman

Dr Derralynn Hughes

Dr Jacqueline Adam

Martynas Davidonis

Plus many more.....



Vilnius Grand Resort, Vilnius

Details & booking form to be sent soon

BE RARE. BE YOU.

RARE DISEASE DAY CAMPAIGN

The Canadian Fabry Association have asked FIN to share their **Be Rare Be You Campaign** with you and your organisation in the hope you will partner with the CFA to help the campaign go global!

The objectives are to get as many people to wear a 'Be Rare. Be You' tattoo around the world to create awareness for Rare Diseases and to sport them on Rare Disease Day. Secondly, to spread empowerment to own your life story and be you as we are all Rare in our own way!

You can join the campaign by:

- Stir up some excitement at your next patient meeting, race, or event and have everyone wearing a Be Rare Be You Tattoo.
- Buy some for your Board Members to wear and share on Rare Disease Day.

Fundraising opportunity:

Interested in a quick and easy fundraising campaign? You and your organization could sell them to university students, interested companies, sports teams, other non-profit organizations, and industry partners!

Here's how it works: The CFA is selling these for $10.00/pack of 2 tattoos and is offering a **discounted rate** to other non- profit organizations at $5.00/pack of 2 tattoos! Thus, creating a fundraising opportunity for your organization to also sell them at $10.00 to supporting sponsors, industry partners, etc..

Help spread awareness for rare diseases, turn it into a fundraiser, share it globally on social media platforms, and join the empowering Be Rare Be You Campaign!

If interested, go to www.fabrycanada.com/shop to place your order

Share Your Tattoo

Share your tattoo widely, as well as tag and post on any of our social media platforms and use the hashtag #BeRareBeYou.

Website: www.fabrycanada.com
Facebook: Canadian Fabry Association
Instagram: Canadian Fabry Association
Twitter: @CdnFabry

BE RARE.
BE YOU.

EVENT: Rare Disease Day – February 28, 2018

Rare Diseases affect 1/12 individuals. Help the CFA launch our Be Rare. Be You campaign by sharing the tattoos with your employer, friends, family, and community to show your support, and empower your soul.

Hearing Loss in Children with Fabry Disease

E. Suntjens; WA Dreschler; J Hess-Erga; R Skrunes; FA Wijburg; GE Linthorst; C Tondel; M Biegstraaten

A paper was published online in the Journal for Inherited Metabolic Diseases this year describing the hearing loss in children with Fabry disease. The authors were prompted to study hearing loss in children with Fabry disease due to the only limited data available on the presence and degree. The Methodology was to collect retrospectively audiogram of the Dutch and Norwegian children with Fabry disease. First hearing sensitivity was determined by studying hearing thresholds at low, high and ultra-high frequencies in children with Fabry disease and comparing them to healthy children.

The results were that 113 audiograms of 47 children with Fabry disease (20 boys with a median age at first audiogram of 12 years were analysed). At baseline slight / mild or moderate to severe hearing loss was present in three children (6.4%, 2 boys). Follow-up measurements showed that three additional children developed hearing loss before the age of 18 years.Of these 6 children, five had sensorineural hearing loss,in the authors opinion most likely caused by Fabry disease.

The conclusion is that a minority of children with Fabry disease show slight / mild or moderate to severe hearing loss, but their hearing thresholds are poorer than the reference values for normal hearing children

J Inherited Metab Dis (2017) 40:725-731

Christine Lavery

News from Fabry NEXT

Fabry Next wants to be your next door neighbour!

Yaeko Ishihara, Organizer

We are a Japanese patient advocacy group supporting people with Fabry and other lysosomal storage diseases.
Our aim is to increase the exchange and sharing of information. Our membership is open to patients, families, health professionals and anyone who is interested in and related to lysosomal storage diseases in any way. Our name, Fabry NEXT, is intended to highlight the name of the disease and to express the meaning of `***go forward to the future***`. The second part was an idea from my doctor. Agreeing a name for our organisation was not easy as some members felt we shouldn't include the disease because there is a sensitivity to having a genetic disease in Japan. However we finally came to agree. In my own experience, it takes a long time to get a diagnosis as the disease is not well known, therefore, using the disease name is essential to create an awareness of Fabry in general society.

Fabry NEXT is your second home
We are very eager to meet with patients and their families whenever possible. We want to talk about their concerns and we want to share in their experiences. We are keen to know more about the diseases affecting the patients and how this impacts their lives. These were the driving reasons for the formation of Fabry NEXT, to be able to offer this contact and sharing opportunities. At the very beginning, we started with a message board via the internet but soon realised this was no replacement for meeting face to face. In 2010, I created a handmade flyer advertising our first meeting in a café and asked my doctor to hand out to the patients with Fabry at his hospital. On the day of the meeting, I was waiting at the café and saw one lady with her perfect smile come over to my table. She was tightly holding my flyer in her hand. I almost cried as I realised that I was not the only one who needed someone to talk to! Since then, Fabry NEXT has reached out to many patients and families in remote areas who wish to communicate with others but it is difficult to travel to Fabry NEXT's home base city, Nagoya.

Fabry NEXT will reach you, no matter how far.
Here some of our activities:

Networking event in Nagano, Aug 6, 2016

Topic: How can we tackle patients' hardship?

..... and in Yamagata, Sept 23, 2017

a lecture given by a cardiologist and followed by a group discussion about genetics and physical conditions that patients are not normally able to talk about to doctors in routine medical care. Useful information and resources for patients were also shared.

Fabry NEXT: Who we are:
We were established in 2012 and our main activities include:

- Networking events and seminars
- Drug manufacturing site and hospital tours
- Participation and booth exhibition at major academic congresses

Scope of operation: Nagoya area and other cities nationwide
Membership: Currently approximately 30 participants
Number of medical advisors: Five
Uniqueness: It is not only Fabry patients who participate in our activities - many patients who have other lysosomal storage diseases as well as their families join us at various events. Our network is expanding nationwide beyond Fabry

Your anxiety is what some-one experienced.
Your concern is what some-one overcame.
We want to help those who need help.
We want to listen to those who need to talk.
Fabry NEXT wants to be beside you to share your feelings.
Today's small courage leads to tomorrow's leap.
Let us hear your voice.

Anyone who wants to share their voice is welcome to write to Fabry NEXT at: info@fabry-next.com

ICIEM, Rio, Brasil

(3) 5-8 September

The 13th International Congress of Metabolic Diseases was held for the first time in Latin America, more specific in Rio, Brasil, Rio. The conference was held in a brand new conference center.

A very extensive and interesting scientific program was prepared, spread over 7 plenary sessions, 16 parallel sessions and 5 pre-congress sessions and 1 breakfast symposium.
In addition, 5 pre-conference satellite symposia and 13 breakfast and lunch symposia were organized by the companies.

About 1000 abstracts were accepted and exhibited in the immense poster space. More than 2000 attendees from 85 different countries were welcomed. This event was made possible by the financial support of 28 sponsoring companies. The morning of September 5th ... it's MetabERN time ... Maurizio gave an update about MetabERN and I gave an update of what the Patient Board of MetabERN has already done and what the planning is..

It would lead me too far to pinpoint all the sessions I attended,... Therefore, it might be better to let me know if you have certain interests and then I can tell you what I know about recent news about it.

For me, it was a nice opportunity to meet again many professors, whom I have known for years .. and I will name and restrict myself to those who have also done a presentation: Prof Giuliani (Fabry), Beck (MPS), Schwarts (Gaucher and PKU), Summar (UCD), Begley (Brains for Brains), Derks (GSD), Eto (Fabry, ..), Kozich (NBS), Kölker (E-IMS and E-HOD), Manoli (MMA) Morava (CDG), Scarpa (LSD), Venditti (MMA).

The most interesting thing about this annual conference is that at this congress you can collect a lot of news, hot from the needle. Many wait till this conference to announce their latest ideas or findings to this outstanding group.

Furthermore, it is also a unique opportunity to negotiate with the top executives of the various companies.

Thanks to my sponsor so that I could attend this highly educative and networking congress !

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Fabry Outcome Survey
Annual Report 2016

Reporting Period: 17-04-2001 to 05-01-2017

Click here for the to access the full report

FOS
FABRY OUTCOME SURVEY

This report has been prepared by Shire Outcome Surveys, on behalf of the FOS Steering Committee

Date of preparation: August 2017
Item code: W-HEOR-0002

News from

OUR GOOD STUFF ™

Being persistently positive in the face of rare disease™

A positive outlook and an optimistic disposition are associated with better mental health and improved quality of life.[1] For people of any age living with a rare disease, and their families, staying positive in the face of immense daily challenges is an important, worthwhile pursuit. But, it's not always easy. A little help can go a long way and being optimistic begins with making positivity a habit.

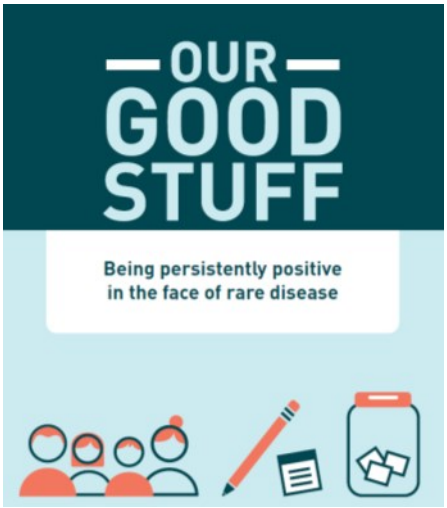
To encourage individuals and families who live with a rare disease, Amicus Therapeutics Patient & Professional Advocacy has created the "Our Good Stuff" kit including a booklet on how the programme works, stickers, notepads and a pen. Encountered in 'real life', this initiative is intended to help people recognise and remember the good things – big and small – in every day, and celebrate them as an individual, family or group. Optimism is contagious and has the power to influence others.

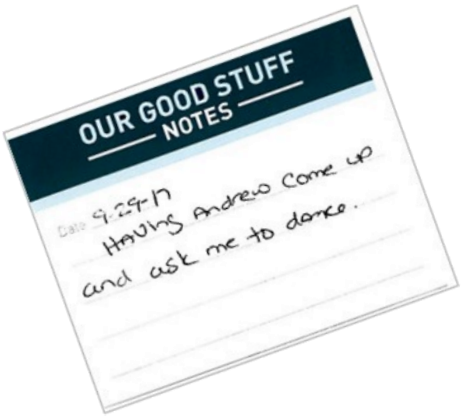
This programme was inspired by the Carter family from the UK, and their experiences of living with health challenges. Recognising their "good stuff" helps them stay positive and motivates them. 'Our Good Stuff' harnesses the Carter family's experience and provides a platform to share it with others, in the hope that even the simplest moments can help lead to inspiring outcomes. For the Carters, optimism has had a remarkable impact on their family – "every year has bad things, but that doesn't make it a bad year", commented mum, Margot Carter.

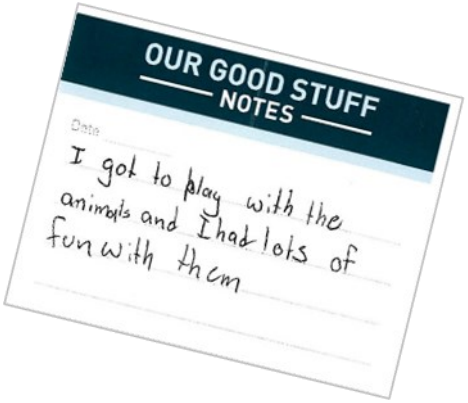
How Our Good Stuff works:

Choose a container to collect your "good stuff." It can be a jar, a box, a bucket or any other container. Use the Our Good Stuff ™ sticker to customise your container and capture your good moments with the paper and pen provided. Then follow these three easy steps:

1. **Reflect.** Record one or more positive moments that happened that day. It can be a happy thought or a big accomplishment.
2. **Collect.** Place the notes into the container, and watch as it fills with "good stuff" over time.
3. **Share.** Read the contents every week or month and celebrate the positive moments!







The Our Good Stuff kit gives people, especially children, who are affected by rare disease, and their families, a tool to develop a more positive outlook. To enrich the experience, Amicus Therapeutics Patient and Professional Advocacy encourages everyone, including patients, their caregivers, their families and their disease communities to get involved and make the most of all the "good stuff" that surrounds us.

Reference:
1. Avvenuti G, Baiardini I, Giardini A. Optimism's explicative role for chronic diseases. *Font Psychol*. 2016;7:295

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