

# Newsletter



## March 2019

### Contents

- FIN Fabry Expert meeting, Spain
- Fabry Women's Day
- Fabry Awareness Month Photo Contest
- Japanese Survey
- News from WORLDSymposium™
- News from FSIG
- News from the Finnish Fabry Association
- Rare Disease Day 2019
- Thank you Toni!

### A word from the President:

*Dear All*



*The first Newsletter of 2019! Spring is upon us and we are very proud to present you with the latest news from FIN!*

*We are finalizing the program for our annual Expert Meeting in Barcelona, we look forward to having you at the meeting! I also want to encourage all of you to participate in our photo-contest we are organizing during Fabry Awareness Month. Please do send us pictures and updates of the activities you are organizing during this month and we will be happy to share this within our network. Of course, you will find a lot of other news which I am sure you will find an interesting read. We also invite you to participate in the Japanese Survey. We would like to emphasise the importance of taking part in this initiative. We would also take a moment to express our gratitude to Toni Ellerton, our coordinator of many years, who will move on to new tasks after the FIN headquarters have moved to the new location in Belgium.*

*Please enjoy reading the FIN newsletter.*

*Looking forward to seeing you in Barcelona!*

*Lut, FIN president*



## FIN FABRY EXPERT MEETING

24<sup>th</sup> – 26<sup>th</sup> May 2019

The primary aim of the Fabry International Network is to facilitate collaboration between Patient Organisations around the world to support those affected by Fabry Disease.



FrontAir Congress Hotel, Barcelona

The Fabry International Network is pleased to announce that the 7th FIN Fabry Expert Meeting will be held at the Front Air Congress Hotel, Barcelona, Spain. Unlike previous meetings we are planning to extend this annual meeting over two and a half days. The programme will open with focus groups on Friday 24<sup>th</sup> May followed on Saturday and Sunday morning by a day and a half of presentations from worldwide professionals specializing in Fabry disease.

We have an exciting program lined up.

Accommodation and events will be at Front Air Congress Hotel, Barcelona, Spain: Which offers a free 10 minute airport transfer from Barcelona-El Prat Airport every 30 minutes. Please see website for more information on airport transfers <https://www.hotelfrontaircongressbarcelona.com>.

Every year we like to encourage as many FIN Member Patient Organisation representatives as possible to participate, but unfortunately have a limited budget and can only offer travel bursary to one person from each organisation

We very much hope that you will be able to attend the FIN Fabry Expert Meeting and should be pleased if you would confirm your place as soon as possible (before April 10<sup>th</sup>) by completing the [booking forms](#) and returning to [info@fabrynetwork.org](mailto:info@fabrynetwork.org) if you have not yet confirmed your attendance.

[Please click here to see the full program](#)\*

\* please note that this program is still subject to change

We look forward to seeing you in Barcelona!

Lut de Baere  
President  
Fabry International Network

## International Fabry Women's Day



Firstly, I would like to introduce our patient association: Fabry Support & Information Group Netherlands (FSIGN) was founded on 9 March 2001 with the aim to unite patients suffering from this condition and look after their interests by giving them a voice.

The FSIGN achieves this by spreading information about the disease and exchanging knowledge regarding the handling and the treatment of the disease. FSIGN works on disease awareness, both for patients, as well as for practitioners and scientists.

The association informs its members through a regular newsletter and the FSIGN website:

<http://www.fabry.nl>.

Annually a general members meeting is organized, where patient information is provided by practitioners and scientists. In addition, the Association assists its members where possible by means of a dedicated patient contact person. Members that start on home infusion are provided with an IV pole. Since 2005 FSIGN also organizes an annual "women's day", every first Saturday of April.

FSIGN is a member of Fabry International Network (FIN), as such our intention is to collaborate with FIN and leverage their existing network, to maximize the outreach Fabry-awareness and to encourage the establishment of a Female Fabry Day.

In 2011 FSIGN won the first PAL-award. This prize was awarded by Sanofi Genzyme. They launched the (PAL) Awards grants program in 2011 to encourage bold new ideas and programs to support the Lysosomal Storage Disorder (LSD) community worldwide. That year, Fabry Support & Information Group Netherlands was the only Fabry organization worldwide that won the prize with a project written by two female board members of FSIGN (both Fabry-affected).

This project was named: Fabry International Female Initiative (FIFI)

An important part was to promote project FIFI internationally by organizing a recurring **International Fabry Female Day.....Every first Saturday of April**

### **Motivation of the Project:**

Women and girls have always been seen as only carriers of Fabry disease. Over the past few years, it has become common knowledge that also women and girls can develop (serious) symptoms as a result of Fabry disease. This is one of the reasons why FSIGN decided to organize an annual women's day.

### **Purpose of this women's day:**

- Peer support contact
- Exchange of experience
- Inventory of needs
- Inventory of problems and ideas that may be of interest to all patients with Fabry disease and other Lysosomal Storage Disorders.

### **Goal of this day:**

To increase recognition and support for all women with Fabry Disease and those directly affected by Fabry Disease (partner, mother, family, friends).

Everyone named above, who is also member of our patient organization, is welcome to join us during the Women's Day. This can also increase your number of memberships!

### **Message:**

In 2013 FSIGN sent the below message by email to all Fabry patient organizations we could find on the internet:

(Since then we received several reactions of organizations that organized an International Fabry Women's Day but we would like to see so much more activities being organized and mark that day on the calendar .....)

**We want to let you know that we, as a Dutch Patient organization, we officially declare the 1st Saturday of April as: International Fabry Women's Day.**

For the past years, the Dutch patient organization (FSIGN) has organized an annual national Fabry women's day for our female Fabry patients and other women and girls directly affected by Fabry Disease. Every year we receive lots of positive feedback for this event. It is an opportunity to come together and share experiences, exchange relevant information, show solidarity and understanding and mostly offer support for each other.

On this International Fabry Women's Day, we want to increase recognition for Fabry Disease and the impact it has on the lives of the women it affects. Our hope is that on this day, activities will be organised for all Fabry women.

It doesn't matter if these are big or modest, physically or mentally challenging activities (depending on the resources available) as everything you do matters.

Here are some examples of the activities we organised in the past for Fabry Women's Day

- A dinner for our female members, where everyone regularly swapped seats to stimulate networking.
- A group dinner to develop an inventory of the most common physical complaints experienced and discuss these together.
- We also organised industry sponsored workshops (don't hesitate to ask for sponsoring to help you get started!).
- A group graffiti workshop: to express emotions creatively and share with each other what Fabry means to them through the form of art.
- A painting workshop to express feelings through painting.
- During our 10th Fabry Women's day, we spoiled our ladies with a beauty treatment and many emotional conversations in between.
- High Tea, with an inspirational speaker.
- Meeting with a storyteller/lyricist and photographer. The lyricist taught us how to write down and express our feelings and sorrows. We took pictures of all participants and together with their stories, being used to create a book about Woman and Fabry disease. (Coming soon)
- Other suggestions: sending postcards, create a telephone circle and organise regional meetings .....

Please let us know how you filled in your own International Fabry Women's Day. We very much look forward to receiving your reports and pictures.

An "International Logo" is developed for this special day.

You can use this logo, to announce your own International Fabry Women's Day activities.

Please let us know if you would like to receive this logo.

Upon request, we can also send you a flyer with the logo, where you can place your own text and use for your activities.



*Erica van de Mheen*  
([treasurer@fabrynetwork.org](mailto:treasurer@fabrynetwork.org))

## **Fabry Awareness Month—Photo Contest**

Each year, during April, we aim to increase the awareness of Fabry Disease by increasing understanding of this rare condition and impact of living with Fabry Disease as a patient and family.

Fabry Awareness Month is all about telling the world about this rare disease. The more people that are aware, the more patients we can help get that early diagnosis. We hope that this year you also join us in sharing details about Fabry Disease with your family, friends and doctors!

**To celebrate Fabry Awareness Month FIN is organising a photo contest!**

### **Fabry & My Future**



Send us a picture during the month of April and let us know how you see your future! Share with us how you make your life valuable every day, despite daily pain or other discomforts. Tell us a bit more about yourself and your picture.

A contest without a prize would not be a contest, so we will choose three pictures and announce the winners at the end of April and make sure the winner receives a nice prize!

Make sure the picture you send in, is a high resolution picture as we plan to print the picture and also present it during our annual Expert Meeting in Barcelona!

Send the picture to: [info@fabrynetwork.org](mailto:info@fabrynetwork.org) before the end of April 2019



## Online Survey from JFA

The Japan Fabry Disease Patients and Family Association (JFA) aims to overcome the disease together with researchers and medical professionals. We also hope to establish the symbiosis society to promote Fabry patients living happily with their own dignity. We wish to be the closest support for spending their everyday life with smiles as much as possible. We planned this survey in hopes that it can help improve the quality of life and improve happiness for Fabry patients and others. We feel extremely honoured that Lut and the entire FIN board have given us this great opportunity.

### **Purpose of the survey**

To improve the lives and treatment balance of patients. We believe that it is very important to know about Fabry patients who live outside of Japan for and how they experience treatments and life as a Fabry patient. We believe this could be the perfect opportunity to collect and exchange information and more importantly learn from each other.

### **Survey content**

The survey investigates all life aspects “Care-Life-Balance” for Fabry patients and families on a global scale . The survey is divided into 6 topics. (About you / Treatment & Study or Work abroad / Employment / About patient support groups / New born screening / Gene Therapy )

### **Survey Respondents**

Any Fabry patients and their family or caregiver(s) regardless of receiving treatment.

### **How to complete the survey:**

The survey can be completed through “Google Form” (available in English only)  
Please click on the link below (or copy and paste this to your browser)

<https://goo.gl/forms/Rtw18DEVfkbAaSKt1>

Answers are anonymous. The data will be collected and used only as a reference point during medical meetings. Your email address and/or any other personal data will not be recorded when you complete this survey.

### **Survey Deadline**

End of Friday March 29th, 2019 GMT

We would greatly appreciate if you could take the time to complete this online survey. Your opinion is extremely valuable to us. A report will be created once all data is collected. The results will be used as only as a reference point during medical meetings. Also, we are very happy to share the results with you at the upcoming Fabry Expert Meeting in Barcelona.

If you have an questions about this survey, please do not hesitate to send a message to:  
[jfaowl2014@gmail.com](mailto:jfaowl2014@gmail.com)

Best regards

Japan Fabry Disease Patients and Family Association (JFA), Hisao Harada, President and the JFA Survey Team

The WORLDSymposium™ is an annual research conference dedicated to lysosomal diseases. WORLD is an acronym for "We're Organizing Research on Lysosomal Diseases". Since its foundation, as a small group of passionate researchers, in 2002, the WORLD Congress has grown into an international research conference that attracts more than 1,600 participants from over 50 countries around the world.

WORLD is not only a great opportunity to connect with our friends and partners around the world, but also an opportunity to look at how we can work more closely with limited resources to improve the lives of the communities we serve.

This congress was again of a high level and a lot of advances were presented there. Not only the speaker sessions were interesting but also the posters were worth seeing.

Science has come a long way, but it is clear that there is still a long way to go in perfecting new treatment options for rare diseases. Gene therapy is again more complicated to get right than we thought. Many initial reports from surveys were somewhat disappointing, but this is going to happen. We have all become too enthusiastic about gene therapy and think this is the answer and it is already perfected. It can be an answer, but many of the technologies need much more work before we can say it for sure. There are other answers, such as new and existing ERTs. We think they will play a major role in the near future and we know that for many people they are a good treatment option. For us, one of the most exciting, also realistic, short-term developments is oral therapies. We know that for some patients they are a good option to give them more freedom, but there are also psychosocial issues that need to be taken into account.



The Pharma industry and science do good things and there are some setbacks, but even these can be considered progress if we learn more about the diseases each time. We understand too well that time is not our friend in the rare disease community, but let us have faith in those who work on new treatments. We have some of the best scientific minds working on new treatments for rare diseases and we are pleased to be part of their dedication to finding a way to create a better future for the rare disease community.

They also paid a lot of attention to clinical trials. Patients participating in clinical trials are heroes and pioneers. The science that is being developed to make these treatments a reality is at the forefront of what is scientifically possible today and we're amazed at how many smart scientists and professionals there are working on these treatments. But without the patients being prepared to participate in clinical trials there would be no new potential treatments.



Participating in a study can be a daunting thought - not just about treatment, but also about how it affects our lives as patients and patient families. We understand the problems that families face when they participate in clinical trials. We know how stressful life can be with a rare disease.

Besides attending this conference, we had many meetings with different companies, who wanted to tell their latest advancements and to make arrangements for cooperation projects, such as the Charity program (Takeda) and the Humanitarian Program (Sanofi). In Belgium it can sometimes be very difficult to get reimbursement for an orphan drug. In some countries there is simply no hope to obtain a therapy or ERT for rare diseases. We, and many others, consider it our moral duty to give hope to these people by working together with industry to provide treatment and therapy for the people in these areas.

Unfortunately, only a small percentage of our rare community can benefit from these programs because it is simply not possible to offer everyone free treatment, but it is a small step in the right direction. These humanitarian programs can only take place through cooperation together with our nonprofit partners from other countries and innovative programs from industry partners such as Sanofi and Takeda. We are proud to be involved, because in the end it is all about you - **rare but not alone**.

Following the WorldSymposium™ there was the annual national meeting of the Fabry Support & Information Group (FSIG). Jack Johnson, Executive Director, FSIG and Vice President for FIN US, invited some of the FIN board members to attend this event.

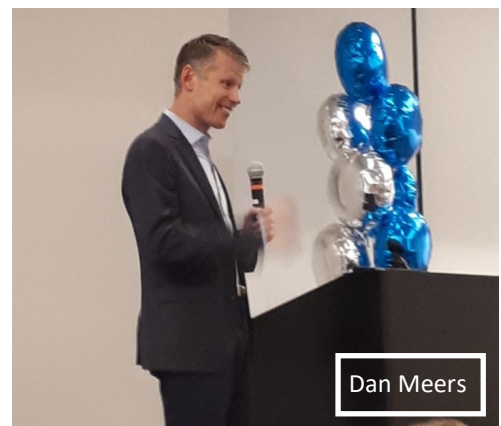
The members' meeting was opened by Jack.

During the meetings, the companies (5) were given the opportunity to give an update to the attendees. In Europe, it is not allowed for companies to have direct contact with patients, but the US has different rules.

There was an update about Fabry disease by Dr. Robert Hopkin. He did this based on the different abstracts and posters that were presented at WORLD, this was a good summary of some of the new information that was presented.

Dr. Lau then gave a presentation about the neurological items and Nadia Ali, PhD presented 'Fabry fog, what is that?' 'Patient empowerment' was again a very interesting topic brought by Dawn Laney, MS, CGC, CCRC.

Day 1 one was a very full day of presentations and we learned a lot. The day ended with a joint dinner, preceded by the welcoming of K.C. Wolf, the mascot of an American football team. During the dinner, Dan Meers gave a presentation about his life after his accident in the stadium. Dan planned to do a bungee jump from a zip line attached to the lighting tower of the stadium as an act at the start of a football game. The accident happened while rehearsing the jump. The jump was partly successful, but he ended up hitting the seats and was catapulted back into the air, and then had to ride the zip line down before being put on the stretcher. He was seriously injured, but recovered after a long rehabilitation. He told the story about his experience in a motivating and humorous way, which inspired many of us. The moral of the story was that people always have choices in life, just by being positive, you can make your life valuable every day, despite daily pain or other discomforts. You should make valuable use of opportunities you are given in life.



The second day was much more informal. Personal stories were told and tips and tricks were exchanged. It even became very emotional occasionally. But it was good to be able to share this with others.

A few examples:

Students can request an adapted program so that they can process the course material at their own pace.

Another example was that some children receive their infusion in the classroom (the nurse stays with them in the classroom), which is not heard of in Europe. One person stated the needle piercing is painless when you stretch or warm the skin. Some patients have a modified infusion schedule (e.g. the full dose every week or every 10 days).



We were very happy to have been part of this meeting.

We noticed that there are many differences between the American and European systems. Especially the fact that there are different types of health insurance policies in America, which means some patient are helped better than other patient. In America everyone can choose their Fabry doctor, but that does not always mean that they know what is happening in this field. Also, the patient is not always heard properly, in other words, doctors do not really listen to the complaints and sometimes refuse to give the best available care or refuse to enroll people in clinical studies, even if they ask for it themselves.



## GREETINGS FROM FINLAND!

The Finnish Fabry Association held its annual meeting this year in the oldest town of Finland called Hämeenlinna close to a medieval Häme castle which was built in the late 13<sup>th</sup> century. The meeting was held in mid-March and there were around 40 participants. This time the lectures covered topics like nutrition for a Fabry patient, living and coping with chronic pain and learning about a new phone app to self-monitor disease symptoms and to fill in the required annual information. The head of the Fabry learning center in Finland, professor Ilkka Kantola, gave a talk about the current situation in Fabry disease in Finland and what can be expected in the future. And yes, there were free time activities as well. From swimming at the hotel spa to outdoor activities and last but not least the insightful discussions between the participants which are always peer support at its best. Laughter and tears, serious conversations and jokes, old members and newcomers, sharing the good and the not-so-good. Those are the things that a successful patient meeting is made of.

*Anna Meriluoto*  
(photos by *Juha Meriluoto*)





## Rare Disease Day 2019

28 February 2019 was the twelfth international Rare Disease Day coordinated by EURORDIS. On and around this day hundreds of patient organisations from countries and regions all over the world held awareness-raising activities. The theme for Rare Disease Day 2019 was 'Bridging health and social care'. Focusing on bridging the gaps in the coordination between medical, social and support services in order to tackle the challenges that people living with a rare disease and their families around the world face every day.

Rare Disease Day 2019 is an opportunity to be part of a global call on policy makers, healthcare professionals, and care services to better coordinate all aspects of care for people living with a rare disease.

Here are some of the amazing activities our members organised:

**Fabry Australia** sold temporary tattoos for Rare Disease Day 'Be Rare.Be YOU'.

They received permission from the Canadian Fabry Association to roll out the campaign in Australia. They started the campaign in Canada in 2018. They are still getting feedback now from members who sold them at schools and work places to raise awareness of Fabry Disease and raise money for Fabry Australia.

One boy living with Fabry Disease read the 'Faber the Dragon' story book which we published to his school friends to explain the condition he and his brother and family members have. His school dressed in plain clothes (no school uniform) to support Rare Disease Day and made donations to Fabry Australia and purchasing the tattoos.



**Rare Diseases South Africa** arranged a Denim Walk at the Walter Sisulu Botanical Gardens on Saturday 23rd February. They had a massive media drive which assisted them in reaching over 19 million people in South Africa on Rare Diseases – please view their media report [here](#). They had a multi-sectoral stakeholder meeting ([more info](#)). They presented at the United Nations on inclusion of rare diseases within the context of UHC ([more info](#)).

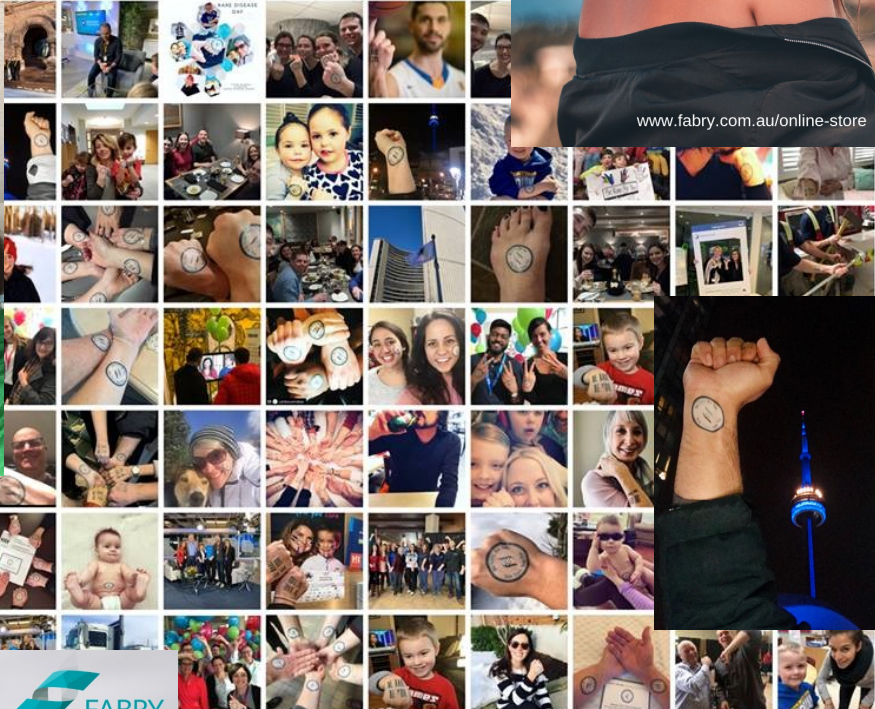
We are all RARE in our own way. The **CFA** set out this Rare Disease Day to raise both awareness and empowerment with their Be Rare Be You Tattoos. The message is to embrace what makes you rare! Their campaign led to lots of media coverage as well as curiosity, discussion, and the magnitude of those who live with rare diseases. Their tattoos reached 16 countries, they sold 1000's, and 2 people even made the tattoo permanent!

Other initiatives:

- Toronto CN Tower lit in blue.
- Nova Scotia, Thornhill, and Toronto all proclaimed Feb. 28th as RDD.



UFA dedicated an entire seminar to Rare Disease Day 2019. Topics such as the diagnose and treatment of the disease were discussed. The seminar was attended by representatives from the Ministry of Health and Public Organisations, doctors, students, patients and their families.





### ***Thank you Toni!***

*The FIN board would like to give their warmest thanks to our coordinator of many years, Toni Ellerton, who will move on to new tasks after the FIN headquarters have moved to the new location in Belgium.*

*Thank you, Toni, for keeping the board organised through several big and small meetings, patiently taking care of most of the background work to help the board and always serving our membership with a smile. You helped the FIN board navigate through a very tough period in its history and always kept us informed about the things that we needed to know.*

*Last but not least we thank you for your ongoing commitment to the field of rare diseases and we wish you all the best in the future.*

*Warmest Regards*

*The FIN Board*

Dear

My name is Charlotte Wauters , and I'm the new appointed coordinator for FIN. It's very exciting to have been given the opportunity to work with the other, very skilled board members of FIN. I'm really looking forward to getting up to speed in this new position—I know I have some big shoes to fill!

I also very much look forward to working with the FIN board to provide help to the Fabry Community.

So, please don't hesitate to reach out with anything you need, I'm happy to help!

Thank you

Kind Regards

Charlotte

[charlotte@fabrynetwork.org](mailto:charlotte@fabrynetwork.org)



Follow [The Fabry International Network](#) on Facebook!

We will be posting regular updates and share the latest information from FIN with you!



[www.fabrynetwork.org](http://www.fabrynetwork.org)  
[info@fabrynetwork.org](mailto:info@fabrynetwork.org)

# Newsletter



## June 2019

### Contents

- Echoes from the FIN Fabry Expert Meeting 2019
- 15th Fabry Women's Day - The Netherlands
- News from the Greek Fabry Association
- News from Avrobio
- Fabry Awareness Month 2019
- FOS Report Takeda
- Fabry Findings
- Impressions from the annual meeting of the Romanian Fabry Organization
- FSIG Newsletter
- "I Hand the Pen To" - Interview with Fabry Leaders around the world

### A word from the President:

*Dear All*



*With the beginning of Summer, we are very proud to present you with the latest news from FIN! This newsletter will serve as good reading materials during your planned vacation!*

*Looking back to the Barcelona FIN Fabry Expert meeting, we are very thankful to have had your presence there and receive your valuable input during the workshops. We received very positive feedback and we will take into account all of your suggestions for future meetings! If you were not able to attend the meeting in person, we have some great news for you! The live stream is still accessible via the links provided in this newsletter.*

*As communicated in Barcelona, included in this newsletter you will find the link to the first issue of Fabry Findings.*

*In order to make this newsletter as informative as possible we are always welcoming news and information from you as (family of) patients, patient organizations and patient representatives. Please send it to our mailbox: [info@fabrynetwork.org](mailto:info@fabrynetwork.org)*

*Please enjoy reading the FIN newsletter and have a great summer!*

*Lut, FIN president*

## FIN FABRY EXPERT MEETING

24<sup>th</sup> – 26<sup>th</sup> May 2019

Continue this very good work! Keep on educating by sharing knowledge to Fabry patients!



Every year the Fabry International Network organizes the Fabry Expert Meeting. This year it was held from May 24th to 26th in Spain – in the capital of Catalonia – beautiful Barcelona and was hosted by the president of the society for Mucopolysaccharide Diseases in Spain, Jordi Cruz, who also gave a presentation about the association he represents. During the meeting we had 78 attendees and among them there were patient organisations' leaders, physicians who are real professionals and have a tremendous expertise level in the field of Fabry disease and representatives of the pharmacy companies. This team led us all to experience interesting presentations, discussions and casual conversations not only during the meetings but also during the dinners.



This year we had two patients speaking – Francisco Albiol who is a Fabry patient from Spain and Wojciech Nadolski – a Fabry patient from Poland. Francisco told us his personal story of Fabry disease and what he had to experience during his journey to diagnosis and treatment. Wojciech guided us through the difficulties they face in Poland where reimbursement for treatment is not provided and this leads many people to develop severe symptoms. He also told us about very interesting initiatives they do to raise awareness about the disease among the government officials.

Doctor Nadia Ali told us about the psychological impact of Fabry disease and about Fabry fog. She also gave us a glimpse to the cognitive function examination procedures and informed that she is coordinating a 12 hour-long cognitive testing for Fabry patients.



Professor Dominique Germain gave a really clear and understandable description of X chromosome inactivation so to say the scientific basis of the Fabry woman. He also gave a presentation about family planning when one of the partners have Fabry Disease.

A representative from the Japanese association presented the results of their survey related to Fabry disease. We were also updated on kidney, heart, pain issues and other interesting topics.

Great experience to learn!

## FIN FABRY EXPERT MEETING

24<sup>th</sup> – 26<sup>th</sup> May 2019

This year's Fabry Expert Meeting also had a new initiative – a day dedicated to workshops. We had topics such as “Understanding the psychological needs of Fabry disease”, “Symptom management”, “Let's talk Fabry men” and “Let's talk Fabry women”. All the patients and their caregivers were divided in to smaller groups to discuss one particular topic and each participant could choose three workshops to participate in. FIN is working on a summary report based on the outcome of the workshops and will share this in the next newsletter. Participant feedback highlighted the desire for continuing the workshop program at future meetings.



As during all past meetings, the annual general meeting of the membership was held to discuss Fabry International Network matters and also re-elected 3 directors of the board for the three-year term. Of course the FIN board and pharmacy companies meetings prior to the Fabry Expert Meeting are to be mentioned. During these we received updates concerning what is happening in the area of treatment and discussed how the industry partners could help us accomplish FIN's mission – “a world where every single person affected by Fabry disease has the best quality of life possible through early diagnosis, treatment and cure”. We will share that information among the membership through our newsletter, website and future meetings.

Thank you for your attendance and valuable contributions to the meeting!

**We look forward to welcoming you in Amsterdam to  
the FIN Fabry Expert Meeting on  
April 24th-26th, 2020!**



## 15th Fabry Women's Day in the Netherlands!

Saturday, April 6th Fabry Support and Information Group Netherlands (FSIGN), celebrated its 15th Fabry Women's Day!!

FSIGN organized a workshop at 'cakes by SUUS' in the exact middle of the country, so everyone was able to attend.. (the longest travel is a two hour drive).

Another location in this town we also use for our Annual Membership Meeting every year.

The workshop 'decorate cakes' was a very big success!



Every woman took a very tasteful cake home with her, to celebrate our special Fabry Women's Day at home with her family.

This special day we celebrate every year on the first Saturday of April and meets a need that we could not imagine...

After all these years we got to know each other better, we have shared each other's joys and sorrows. We can comfort each other after bad results and support each other in difficult decisions. This is invaluable!

I would like to share a few experiences of participants:

*"What a great day it was. The 'cakes by SUUS' were delicious and so good to see you all again, it really has something special, it feels a bit like family... So much fun to talk with each other again and see how it goes. After the workshop we went for dinner. Was delicious and cosy. We sat at a nice table with a new member, so fun and very clever! When we are together, we feel strong!"*

*"Have you missed this party or are you still unsure if you should come?  
I would say: do it, the more the merrier...."*

*"We love to be every now and then, in a company of people who, like no other, understand what you're talking about if you tell about how Fabry takes up space in your life. And after all these years of membership, those people are just a little 'family'. We think it's very nice to see and speak to each other now and then. And so we met again, last April 6th, cosy with a large group of chatting ladies at 'cakes by SUUS' in Almere, on the annual international women's day!"*



As one of the two female board members of FSIGN, I hope that other patient organizations are also able to bring together women involved with Fabry, as there are Fabry-affected, mothers of Fabry-affected, wives of Fabry-affected, daughters of Fabry-affected, sisters of Fabry-affected, friends of Fabry-affected, so we can support each other!

Our only stipulation is that you need to become a member of our patient organization...

This is one of the reasons our patient organization in our small country (17 million inhabitants) has almost 150 members, of which 53% are Fabry-affected.

Good luck with organizing your own International Fabry Women's Day, April 4, 2020!

Erica van de Mheen

## First Fabry Patient meeting ever in Athens, Greece



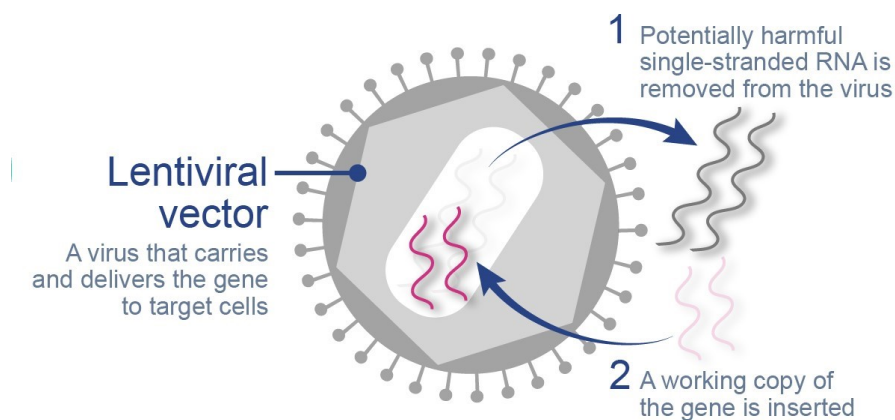
On April 21st, 2019 the very first Fabry Patient Meeting ever was held in Athens, Greece. The meeting took place in Myrtillo café inside the beautiful Kapaps Park. Myrtillo café is a Social Cooperative Enterprise with a Special Purpose; the integration of vulnerable groups into the labor market. It was the ideal place to celebrate vulnerable and special people like Fabry patients during the Fabry Awareness Month. The Board of the Pan-Hellenic Association of Lysosomal diseases "Solidarity" organized a special gathering for Fabry patients. It was an opportunity to discuss and exchange experiences, dreams and fears. The atmosphere was lively and friendly. First the President, Kate Theohari, did an introduction and then the Vice President of the association and Secretary of FIN, Mary Pavlou, did a short presentation about Fabry Disease, the history of the association and we also talked about the new and emerging treatments for Fabry. Afterwards, coffee and a light lunch was in order, talking again about dreams and new adventures. Pictures were taken to create beautiful memories of the meeting. It was for sure a good first step to communicate better with our members and also to find ways to overcome the burden of the disease. We promise to organize it again next year!

*Mary Pavlou*



AVROBIO, a leader in lentiviral-based gene therapies, is a clinical stage company developing disruptive therapies that have the potential to transform patients' lives in a single dose.

Gene therapy involves the transplantation of normal genes into cells in order to correct genetic disorders. Gene therapy uses a vector (see illustration below) to carry and deliver a working copy of a gene to the body's cells. A vector is a deactivated virus that is unable to reproduce and spread (#1 below). There are different types of vectors. AVROBIO's **Plato™** is a gene therapy platform which uses a lentiviral vector and is designed to target underlying genetic disease by restoring normal gene function (#2 below) and enzyme production.



For more information about AVROBIO's pipeline and technology please visit <https://www.avrobio.com/>.

## Fabry Awareness Month 2019

Each year, during April, we aim to increase the awareness of Fabry Disease by increasing understanding of this rare condition and impact of living with Fabry Disease as a patient and family.

Fabry Awareness Month is all about telling the world about this rare disease. The more people that are aware, the more patients we can help get that early diagnosis. We hope that this year you also join us in sharing details about Fabry Disease with your family, friends and doctors!

Here are some of the events that were held by our members during this month:



Fabry Canada is continuing to travel across Canada holding their Patient Empowerment Meetings and bringing both awareness and education. Through the month of April they shared patient stories, gene therapy experiences, and a female perspective with media across the country.

MPS Turk published this [video](#) on their social media.

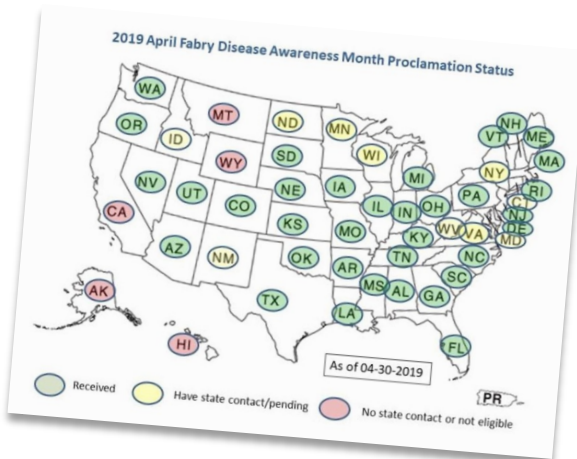
The U.S. National Fabry Disease Foundation (NFDF) coordinated with Fabry community members across the country to request state governor's proclamations for April 2019 Fabry Disease Awareness Month. Of the 50 states in the U.S., 35 state governor's proclamations were received this year as highlighted in green on the map.

The states highlighted in yellow on the map are states where proclamations were possible but attempts this year were unsuccessful for various reasons. The states highlighted in red have requirements that are difficult to meet or the state does not participate in a governor's proclamation program. Each year the NFDF and Fabry community state representatives work together to obtain as many proclamations as possible to maximize Fabry disease awareness.

In addition to obtaining formal governor's proclamations, the NFDF published a daily Fabry disease awareness post on their face book page at [www.facebook.com/FabryDisease](http://www.facebook.com/FabryDisease). The daily posts addressed a wide range of Fabry disease awareness topics to help people better understand, manage, and cope with Fabry disease and to better understand Fabry community programs.

The number of people who viewed the daily awareness posts in April ranged from about 1,300 to over 11,000. The number of people who follow the NFDF face book page is over 3,200 to date and growing.

You can see photos of many governor's proclamations and participating community members in the "2019 Fabry Disease Awareness Month Photo Album" at [www.facebook.com/FabryDisease](http://www.facebook.com/FabryDisease).



The 2019 Chinese Fabry Patient Conference, cosponsored by the Shanghai Four leaf Herb Family Care Center (formerly known as the Chinese Organization of Rare Disease , CORD) and the Chinese Fabry Patient Community , was successfully held in Beijing. Nearly 100 people including Fabry experts, representatives of pharmaceutical companies, rare disease genetic testing institutions, social welfare organizations, patient organizations and patient groups attended.

In this conference, the Fabry Patient Community released the status of the domestic Fabry group. Many patients shared their stories living with Fabry. Experts shared the diagnosis and treatment of Fabry Disease and the status of Fabry drugs. An intense discussion was carried out at the conference. During the meeting, Beijing 301 Hospital, Beijing Dongzhimen Hospital, Beijing Anzhen Hospital, and Beijing Tongren Hospital also conducted a free onsite clinic for the Fabry patients who participated in the conference.

Welcome to the 2017 Fabry Outcome Survey (FOS) Annual Report, which provides an overview of FOS as of 5 January 2018. This report includes details of the FOS Steering Committee as well as a summary of patient demographics and the publications that have been developed using the data collected in the registry. FOS (ClinicalTrials.gov identifier: NCT03289065) is a large, global, multicentre, observational registry, sponsored by Shire, for patients with Fabry disease. The registry was established in 2001 with the aim of collecting real-world data on the long-term safety and effectiveness of enzyme replacement therapy (ERT) with agalsidase alfa and the natural history of the disease. FOS became a disease registry during 2017 and is now enrolling any patient with Fabry disease, including those who have received an approved treatment for Fabry disease other than agalsidase alfa.

### Key highlights from FOS, as of January 2018

- As of January 2018, there were 3515 patients enrolled in FOS, which represents an increase of 11% compared with January 2017. In total, data have been collected by 137 centres in 25 countries.
- Similarly to previous years, 57% of enrolled patients are female and 12% are children (defined as < 18 years old at FOS entry).
- Overall, 2171 patients (62%) have received one or more doses of approved treatment for Fabry disease: agalsidase alfa (n = 2112), agalsidase beta (n = 63) or migalastat (n = 34); 38 patients have received more than one approved treatment. The relative proportions of patients who have received agalsidase beta or migalastat are lower than would be expected based on worldwide use of Fabry treatments because enrolment of patients receiving approved treatment for Fabry disease other than agalsidase alfa began in 2017.
- Four manuscripts based on FOS data were published during 2017, so that there was a total of 54 FOS publications as of January 2018. In addition, three posters based on FOS data were presented at two meetings in North America and South America.

Takeda would like to take this opportunity to thank all of the patients and their families, and the physicians and their staff, who have participated in FOS and contributed data to the registry.

[Click here](#) for the full report

A decorative, light grey, curved line that starts from the left and curves upwards towards the right, positioned below the 'Click here' link.

## FABRY FINDINGS

Scientific publications that are translated into lay language made available to the Fabry Community;  
A new article will be announced in every FIN newsletter.

The first article is based on the publication:

*Predictors of objective cognitive impairment and subjective cognitive complaints in patients with Fabry disease*

Simon Körver 1, Gert J. Geurtsen<sup>2</sup>, Carla E. M. Hollak<sup>1</sup>, Ivo N. van Schaik<sup>3</sup>, Maria G.

SCIENTIFIC REPORTS (2019) 9:188 DOI:10.1038/s41598-018-37320-0

### Fabry Findings

Issue No. 1 | Summer 2019



#### What is Fabry?

Fabry disease is a rare, genetic condition which is estimated to affect around 1 in 100,000 people.<sup>1</sup>

In Fabry, an absence or reduced level of an enzyme called  $\alpha$ -galactosidase A ( $\alpha$ -Gal A), means that the body cannot break down certain types of fats, called globotriaosylceramide (GL-3) and plasma globotriaosylsphingosine (lyso-Gb3), and GL-3 builds up in a variety of cells in the body.<sup>1</sup>

This build-up causes damage to tissues and organs and leads to a range of symptoms and complications, which vary from one person to another.<sup>1</sup>

Disease progression is influenced by the sex of the individual (male or female) and how the disease presents, called its **phenotype**, which is classified as either **non-classical** (mild form) or **classical** (severe form).

Symptoms and complications vary from one person to another<sup>1</sup>

|  |  |  |
|--|--|--|
| <b>SKIN</b>  | <b>KIDNEYS</b>   | <b>EYES AND EARS</b>   |
| <ul style="list-style-type: none"><li>Sweating less than normal</li><li>Small dark red/purple spots located between the belly button and the knees</li></ul>   | <ul style="list-style-type: none"><li>Protein in urine</li><li>Decreased kidney function</li><li>Kidney failure</li></ul>  | <ul style="list-style-type: none"><li>Hearing loss (in children)</li><li> ringing in ears</li><li>Cloudy vision (cataracts)</li></ul>              |
| <b>BRAIN AND NERVES</b>  | <b>STOMACH AND BOWELS</b>  | <b>HEART</b>   |
| <ul style="list-style-type: none"><li>Burning in the hands and feet</li><li>Intolerance to heat/cold</li><li>Vertigo/feeling dizzy</li><li>Pain</li><li>White matter lesions</li><li>Depression</li><li>Mini stroke (TIA)</li><li>Stroke</li></ul> | <ul style="list-style-type: none"><li>Feeling sick/being sick</li><li>Diarrhoea</li><li>Pain/bloating after eating</li><li>Difficulty managing weight</li><li>Feeling full after eating a small amount of food</li></ul> | <ul style="list-style-type: none"><li>Irregular heart beat</li><li>Enlarged heart</li><li>Heart attack</li><li>Heart failure</li></ul>             |
|  |  | <b>OTHER</b>   |
|  |  | <ul style="list-style-type: none"><li>Tiredness that is not relieved by rest or sleep</li><li>Shortness of breath</li><li>Cough/wheezing</li></ul> |

[Click here](#) to read the first issue of Fabry Findings

The Fabry Findings articles are published on the FIN website

<http://www.fabrynetwork.org/fabry-findings/>

We encourage you to translate  
the articles into your own language so these can be shared in your community.

## Annual meeting of Romanian Fabry Organization



This year, the annual meeting of the Romanian Fabry Organization took place on June 5th and 6th in Bucharest. The meeting started with a press conference.

Alexandru Tcaciuc, President of the APBFR association proudly represented the organization. Prof. Ruxandra Jurcut, a cardiologist who follows up Fabry patients in Bucharest with a multidisciplinary team briefly explained the course of the disease. Dorica Dan, Chairman of the Prader Willi PO, Chairman of the national rare disease organization RONARD and Eurordis board member emphasized the importance of networking as a national PO and joining European and international umbrella organizations. Lut de Baere, the day to day operations of MetabERN and FIN were highlighted by her.

According to the calculations, theoretically around 200 Fabry patients are living in Romania. APBFR was established only recently, namely in 2018. They have 3 directors and 26 members. They have a website, an open and closed Facebook group and work intensively with RONARD, various LSD PO's, Prof. Jurcut (center of expertise), government services and Sanofi Genzyme. In Romania, only Fabrazyme is being reimbursed. During the lecture, a promotional film was shown in which various Fabry patients spoke about their experience with Fabry Disease.

APBFR supports its members in daily life. For example, they mediate when patients experience problems, if the therapy is not given on time or if the dose is not adjusted. Some patients receive the diagnosis, but cannot accept it, deny the severity of the disease and sometimes refuse treatment. It is up to Alexandru to point out to these people that treatment will more or less stabilize the disease and that the treatment will improve their quality of life. Alexandru also addressed the members to support the organization so that the workload can be divided.

The expertise center of Prof Jurcut was founded in 2015. She follows 9 Fabry patients. A total of 25 Fabry patients are being treated currently and 34 have been diagnosed. Fabry patients have an annual check up. They can always reach out to Prof. Jurcut, through the closed Facebook group. The patient is central to the care and a lot of attention is paid to the immediate environment and the caregivers of the patient. The center is responsible for the education of doctors and paramedics in local hospitals.

Of course genetics could not be missing from the program, this was brought by Dr. Viorica Radoi. Prof. Jurcut expertly addressed the problems with the heart. Dr. Elena Rusu was the one who told the attendees more about the functioning of the kidneys. The neurological aspects were examined by Dr. Adriana Dulamea. How the treatment of the children works was explained by Dr. Cristina Stoica. NKO vertigo. Deafness is a common problem and has been clarified by Dr. Madalina Georgescu. The different available treatment options were discussed by Dr. Lucia Ciubotaru

On day two the newcomers could watch the introduction of the association and the promotional video. Afterwards they took part in the given lectures. 'Pain treatment' was brought in a very clear way by Dr. Vlad Stefanescu. The topic 'Depression' was brought by Dr. Maria Radu. She told in a very intelligible way how one can get stuck in a vicious circle. Most patients suffer from chronic pain or pain at regular intervals. Because of that pain you easily end up in this circle, but it can be broken. She offered those present various options: medication, but also other methods that can lead to breaking this circle, namely positive thoughts, holding on to beautiful moments in life, falling in love, etc.

A separate session was given for nurses to teach them how to correctly administer the ERT.

The psychological impact when the diagnosis is communicated to the patient followed by group therapy for the patients and their caregivers was steered in the right direction by Elena Marinescu

And so this super busy but very fascinating two-day event came to an end.

My sincere thanks to Alexandru for allowing me to get to know him and his PO better.

Wishing him all the best for the association and keep up the good spirit!

*Lut De Baere*





Did you get a chance to read the FSIG Newsletter?

[Click here](#) to read it.



### Live Stream FIN Fabry Expert meeting

If you were not able to attend the FIN Fabry Expert Meeting in person, we have some great news for you!

The live stream is still accessible via the links here below

Saturday, 25th May: <https://youtu.be/czxSFBnREBg>

Sunday, 26th May: [https://youtu.be/HO0ihJsBn\\_I](https://youtu.be/HO0ihJsBn_I)

## FOCUS ON FABRY LEADERS AROUND THE WORLD

### Interview with Megan Fookes from Fabry Australia



**When did you join your national patient association?**

1994 – when it formed and I am actively involved from 1999.

**What was the reason for joining?**

*My father, uncle and I had Fabry Disease and I saw the importance of forming a Fabry disease community to support one another and raise awareness and funds for research into Fabry disease.*

**What are the vision and mission of your association?**

*Fabry Australia's Mission is Uniting the Australian Fabry Community, by working together to improve the lives of those affected by Fabry disease*

*Fabry Australia's goals are:*

1. *Promote Patient Advocacy*
2. *Share information, knowledge and resources*
3. *Promote and support research and development*
4. *Encourage active involvement with patients, clinicians, medical personnel, partners and industry groups*
5. *Raise awareness and understanding of Fabry within the Community*
6. *Work co-operatively with other Australian and international organisations to promote best practice support and lifelong care for people diagnosed with Fabry disease*
7. *Maintain financial and resource viability*
8. *The Business Strategy is drafted and reviewed by the Management Committee of Fabry Australia every 2 years and it is shared with the members and supporters on the Fabry Australia website.*

**How many members are there?**

*Fabry Australia has over 300 Fabry patients diagnosed. Fabry Australia has 768 followers on its Facebook page and 226 members on its Fabry Australia Discussion Page.*

**What do you consider to be the major achievements or activities that you are proud of?**

*Fabry Australia is very proud of its advocacy in Australia managing to get both Enzyme Replacement Therapies (Replagal and Fabrazyme) listed on the Life Saving Drugs Program (LSDP) of the Australian Department of Health for reimbursed therapy in 2004. This was with the help of the many members, supporters of Fabry Australia who each wrote to their local Federal politicians asking for their support to list the treatments on the LSDP and it was done before computers were readily accessible.*

*Fabry Australia is proud of the first Fabry Clinic which was set up in 1994 in Melbourne and has been replicated around the country. There are now 5 adult clinics and 5 paediatric clinics across the country managing 300 Fabry patients.*

*Fabry Australia is proud of its online presence with its website which is accessed by many Fabry affected across the country and the world. We are also very proud of our strong social media presence which has helped us grow and reach members across the country and around the world.*

*Fabry Australia is proud that it is turning 25 this year and is still going strong.*

*Fabry Australia is proud to advocate for positive changes to the LSDP with a new listing of Galafold Migalastat for Fabry patients in Australia.*

*Fabry Australia is also proud it has established a Medical Advisory Committee who have helped strengthen and support the advocacy work of Fabry Australia to ensure people diagnosed with Fabry disease can access quality health care services and access to new clinical trials/research.*

*Fabry Australia is proud of its ability to host bi-annual Fabry Expert Conferences in Australia to ensure its members are up to date with the latest information and developments in Fabry Disease research as well as tips/tricks to managing their disease with other support services from allied health and social care services.*



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## FOCUS ON FABRY LEADERS AROUND THE WORLD

### Interview with Megan Fookes from Fabry Australia

#### Can you name some challenges that your association is currently facing?

*Some of the challenges Fabry Australia faces is ensuring the Clinical care and management of Fabry patients is well supported. We would like to see accreditation be given to Fabry Disease Centres of Excellence that offer 'in house' multi-disciplinary clinical services and access to ongoing clinical trials and research. Fabry Australia would like our Government to fund such services and ensure there is incentives to strengthen and grow such services. The demand on the clinics is increasing as more patients are diagnosed or need support. Fabry patients are living longer and this puts more stress and strain on health care services that currently are stretched beyond their capabilities.*

*We are concerned that we need more Fabry Doctors to train and understand the condition. As the current experts retire, we want to know their knowledge and expertise is passed down to new Fabry doctors who will continue the good work.*

*Access to reimbursed therapies is challenging in the absence of a Rare Disease Strategy and policy in Australia. The criteria for receiving funded ERT's or Galafold is strict and needs urgent review. Fabry Australia is concerned that Fabry patients are accessing delayed treatment due to the restrictive criteria. In Australia, a patient must receive 12 months' ERT before being offered Galafold which is unlike the rest of the world. Fabry Australia would like to see this clause adjusted and removed from the criteria.*

*In Australia patients receiving ERT are monitored by the clinics each year and all the results and data is sent to the Government (LSDP). This is a lot of paperwork for the clinics and seems unnecessary for Governments to be receiving. Fabry Australia would like to see processes changed to ease the burden to patients/clinics and encourage more patients to receive earlier access to treatments.*

*In Australia, it is very difficult to receive treatment for Fabry disease as a paediatric and typically it is delayed. Again – Fabry Australia would like to the criteria reviewed to be more in line with what the rest of the world is doing with treating children diagnosed with Fabry Disease.*

#### Can you name some future goals or plans?



*Fabry Australia is hosting its 25th Anniversary Silver Dinner at its Fabry Conference in October this year. This is a huge milestone.*

*Fabry Australia is planning to host its 3rd Fabry Retreat in 2020 and hopes to bring this to Queensland. Other future plans and goals will be mapped out at the Management Committee's Strategy Day in October this year. We will be mapping out the next 5 years of goals and plans at the review of the strategy with a facilitator and can share more after this.*

#### Has your association you had issues with the national health system or insurance problems?

*Yes. Recently – with the listing of Galafold. This took a very long time to get listed with a rejection initially and also with the clause in the criteria that patients must receive 12 months ERT before they can receive Galafold.*

#### How would you describe the current treatment situation in your country?

*As mentioned earlier, the current treatment situation in Australia is a bit restrictive. Of the 300 Fabry patients diagnosed only one third are on treatment. Typically, children do not receive treatment or around the age of 15/16 they may access it but it is difficult to get.*

#### What are the major issues with Fabry disease in your country?

*Access to treatment can prove difficult if the patient doesn't meet criteria. If you live in rural/country areas, it is a long distance to the treatment centre. In Australia the first 12 infusions must be done in the major Fabry Clinic and for some this can be up to 7 or more hours away. Receiving ERT at home is difficult for some patients who live in remote country/ rural areas. The home infusion nurses can't get to those locations.*

*Fabry patients must do a lot of tests each year which are sent to the LSDP (Department of Health in Canberra) by May each year to ensure ongoing access to treatment.*

*Some Fabry Clinics can manage to co-ordinate clinical care/ appointments / tests at the Clinics and others, send patients away to other centres. This proves difficult for Fabry patients as this is more time off work / school to attend more appointments and fortnightly infusions. Some patients have lost jobs due to too much time off work.*

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# FOCUS ON FABRY LEADERS AROUND THE WORLD

## Interview with Megan Fookes from Fabry Australia

### How would you further raise global awareness for Fabry disease?

*Fabry Australia collaborated with the Canadian Fabry Association and its 'Be Rare. Be YOU' temporary tattoo campaign this year for Rare Disease Day and Fabry Awareness Month. We would like to see this rolled out globally and do this again next year. We would like FIN to promote the campaign and share with other Fabry Patient Groups globally to get behind the campaign to raise awareness of Fabry Disease as part of the rare disease movement and during April Fabry Awareness Month as part of FIN's social media strategy.*



### What are your suggestions for future projects for FIN?

*Fabry Australia would like FIN to do work on the need for a single Fabry Disease Registry (owned by the patient body, independent of industry). Currently there are multiple registries (but these don't have patients not on treatment listed on them).*

*Access to treatment. Fabry Australia would like to do work on this with other groups to see what criteria is in place in other countries to access funded therapy. Doing some work on this face to face is important and would help each of our groups globally as we advocate to see reimbursement strengthened. Fabry Australia would like to see global consensus on the criteria to access therapy and clinical management and care of patients. There is Delphi Project being drafted and it would be great to review this when published and have input from the leaders of Fabry patient groups globally to ascertain improved clinical care and management of Fabry patients.*

*Apart from the annual conference, Fabry Australia would like to see FIN active in other projects that make a real difference to the Fabry patient groups who are supporting Fabry patients. Looking at positive patient empowerment mindfulness approaches to self-management of Fabry disease. There is a lot of focus on treatment / disease itself, but not on the whole patient. Building upon the psycho-social side to Fabry disease, building some useful tools to support Fabry patients would be practical and helpful to patients and the groups supporting them.*

### What would you like to explain to other people / doctors / nurses / decisionmakers about Fabry disease?

*Fabry disease is invisible and can't be judged on first appearances of a person diagnosed with it. Fabry patients often suffer in silence and do find day to day very tough. Fabry pain can be very debilitating and ongoing symptoms of Fabry disease is a challenge. There is a need to make life a little easier for those living with Fabry disease. Often a patient who has Fabry disease, may have experienced long delays in obtaining their diagnosis and have been through the healthcare system with not a lot of support. There is a need to ensure Fabry patients are listened to, heard, correctly managed and can access best possible care/treatment and access to new clinical trials/ contributing to research with no delay.*

### What kind of information are we still missing about Fabry disease?

*The impact Fabry disease has on the patient, their families and their communities. This is starting to be discussed, but there is a need to do more work on this. There is a need to develop educational tools that support this further (independent of industry and branded by FIN). Having an educational resource / tool to show schools or a similar tool to share with work place is important to communicating a bit more about the condition in simple terms and how it impacts a child or an adult.*

### What is your most effective means of taking your mind off Fabry disease?

*Having a bath. Spending time with my family / friends. Sitting under a blanket and watching a good TV show/ movie. Laughing! Yoga.*

### Is there anything else you would like to add?

*Thank you for the opportunity to share some thoughts and ideas. It would be great to have all the groups do this collectively so we can bounce ideas off one another and collaborate for the global Fabry community.*

**Thank you for your time Megan!**



**Megan hands the pen to Mikhail Kuznetsov -  
The Road To Life, Russia**



#### **Fabry International Network**

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Charity Registration Number 04080030 The Chamber of Commerce Drenthe The Netherlands



# Newsletter



## October 2019

### Contents

- Save the Date Expert Meeting 2020
- News from SSIEM 2019
- A closer look to Gene Therapy
- News from Poland
- Fabry Awareness Month 2019 Winner
- Summary Reports Workshops Expert Meetings
- The Rare Fair
- "I Hand the Pen To" - Interview with Fabry Leaders around the world
- Publication: Results from an International Patient Survey

### A word from the President:

*Dear All*



*We hope your summer was as great as ours was!*

*After relaxing holidays we are now back on track to continue to raise awareness about Fabry Disease! At the beginning of September FIN attended SSIEM in Rotterdam and we also presented our very first FIN booth. We met with colleagues, pharmaceutical partners and attended lots of interesting sessions.*

*The Board is delighted to share with you some very good news from Poland!*

*The situation in Poland has been addressed in many of our past newsletters and it'll be very exciting for all readers to see that they finally have the reimbursement issue solved. Read the full story on page 6.*

*We are also very happy to share with you the outcome of the workshops that were held during the Expert meeting in Barcelona. At the moment we are already focusing on the organization and program for the 2020 meeting! We will be sharing more information soon!*

*In order to make this newsletter as informative as possible we are always welcoming news and information from you as (family of) patients, patient organizations and patient representatives. Please send it to our mailbox: [info@fabrynetwork.org](mailto:info@fabrynetwork.org)*

*Please enjoy reading the FIN newsletter!*

*Lut, FIN president*



# **FIN FABRY EXPERT MEETING 2020**

## **SAVE THE DATE**

**We look forward to welcoming you in Amsterdam to  
the FIN Fabry Expert Meeting on April 24th-26th, 2020!**

**The meeting will take place at the [Steigenberger Hotel](#) near Schiphol Amsterdam**

## **Preliminary program**

**Thursday April 23rd: arrivals**

**Friday April 24th: workshops and AGM followed by dinner**

**Saturday April 25th: Expert Meeting followed by dinner**

**Sunday April 26th: Expert Meeting ending at 3pm**

**We will be sharing more information soon!**

## A glance of the annual symposium 2019 of the Society for the Study of Inborn Errors of Metabolism



Fabry International Network's vision is of a world where every single person affected by Fabry disease has the best quality of life possible through early diagnosis, treatment and cure. This can be done by organizing and helping to organize various initiatives, Expert Meetings and publishing a quarterly newsletter, also getting into direct contact with other people from the Fabry disease community: patients, family members, caregivers and other stakeholders. We also see gathering knowledge in order to provide the latest information to the membership as a crucial activity for our board. Therefore we attend scientific conferences related to Fabry disease and this time I want to give you a glance of the SSIEM (stands for - Society for the Study of Inborn Errors of Metabolism) annual symposium 2019 in Rotterdam, the Netherlands.



President Lut de Baere and four members of the board – Erica van de Mheen, Anna Meriluoto, Mary Pavlou and Martynas Davidonis attended the congress. We were very happy to present the FIN booth at SSIEM. For us this was a very successful event where we met with several new and known people to speak about FIN and its mission and how we could support each other. We are very grateful for this opportunity and will continue to share information through congresses such as SSIEM as FIN aims to facilitate, support and enable the Fabry community.

We also attended lectures and I might say that the future for all patients with inborn errors of metabolism as well as for Fabry patients looks very promising and science is moving forward quickly. Nowadays we might start looking forward to finally having a cure (what was just a theoretical possibility 20 years ago). The speakers at SSIEM were very strong and gave wonderful insights of what they do every day to work on a better quality of life for patients. Some of the lectures were easy to comprehend and some required specific knowledge or a medical background.

There were very few lectures about Fabry disease but there were more than 10 of them which dealt with the matters of novel treatments in general and keeping in mind that these diseases at some point have similar mechanisms this might be applied for Fabry disease at a later stage.



The FIN board members had chance meet with the pharmaceutical partners: Takeda, Sanofi-Genzyme and Amicus Therapeutics where we were informed about what is happening regarding the Fabry disease treatment and we also discussed our future collaborations. We also took the opportunity to have a board meeting after the sessions and meetings where we could discuss important issues that needed to be addressed.



Wrapping up this article I would like to express my happiness about the new medication which will hopefully be registered and could be used for multiple diseases as one of them is Fabry disease! Here we see a new concept of multi-disease treatment. Stay tuned for more details and news in the near future!

Martynas Davidonis





## A Closer Look at Gene Therapy

Our genes make us unique. Inherited from our parents, genes help determine physical features and traits like height, eye and hair color. Genes are made up of DNA, which are genetic instructions to build enzymes and proteins. Enzymes and proteins make muscles, bones, and blood, among many other things in the body, which support most of the body's functions, such as digestion and transporting oxygen through the blood.

Genes that are missing from birth, or don't work properly, can cause disease because the proteins built from the genes are missing or defective. Scientists have been working for decades on ways to modify genes or replace faulty genes with healthy ones to treat, cure or prevent a disease or medical condition.

[Click here](#) to read the full article on Gene Therapy on the FIN website

## Association of Families with Fabry Disease in Poland - Treatment of Fabry disease in Poland is finally reimbursed!



The history of Association of Families with Fabry Disease in Poland starts 17 years ago with two families, that initiated and introduced into life the idea of bringing all Polish Fabry patients together as one big unit, not just single individuals. It is well known that there is strength in numbers and if you want to be heard you need to make others listen. Well, one of these families was my own so the association is something that I grew up with.

Since the beginning, the idea was to help all Fabry patients to get a proper medical care and treatment that would stop the disease. At that time, there was no treatment available in Poland. However, there were clinical trials opened in Poland which in the end, gave a chance for a decent life for at least 20 patients. Unfortunately, once the clinical trials were closed, no one else had a chance for the treatment. It became clear that we needed support from the Polish government and the Ministry of Health if we wanted to help the rest of patients. We didn't know how difficult the path we've chosen really was. With really basic knowledge about regulations, having no experience with the political and medical world, no contacts and no connections at all, we were doomed to fail. But, we didn't give up.

Time flew by, the number of Fabry patients grew, our Association got bigger and bigger. But there were many people who suffered from the disease with no medical support. We were more than aware of the fact that knowledge about rare diseases are still too little to treat these people properly. Moreover, we were also aware of the fact, that without reimbursement of the treatment, doctors were basically powerless. It was also clear that as such a small group we are invisible, so in 2007 we became a part of National Forum for Rare Disease "Orphan". Also, we got connected with Fabry International Network.

Luckily for us, in 2013, a pharmaceutical company applied for reimbursement for Fabry disease treatment. The process was long, difficult and stressful. The Association tried as much as possible to show how important reimbursement for Fabry treatment is, so it was completely devastating when the final decision of Ministry of Health was negative. A big question mark appeared on the Fabry patient's lives.

At that time, only about 20 people were treated with the enzyme replacement therapy in the charity programmes sponsored by pharmaceutical companies. Still, a number of patients were left without proper care and treatment, suffered in their own homes. The feeling of disappointment after failure of the reimbursement process changed into furiosity, and furiosity evolved into motivation. Still, without support from pharmaceutical companies our hands were tied, because according to the local law, only a manufacturer of the treatment can apply for reimbursement.

After a negative decision in the first process, we were afraid that no one would ever try again. It took 4 years but it was definitely worth waiting, as not one but two companies submitted their applications for reimbursement of Fabry therapies. We assumed that such competition might be beneficial for the entire process. Not much later the third company joined the process. The feeling that this was our 'now or never' moment was in the air. With the help of external companies, the Association started to make noise in the local media.



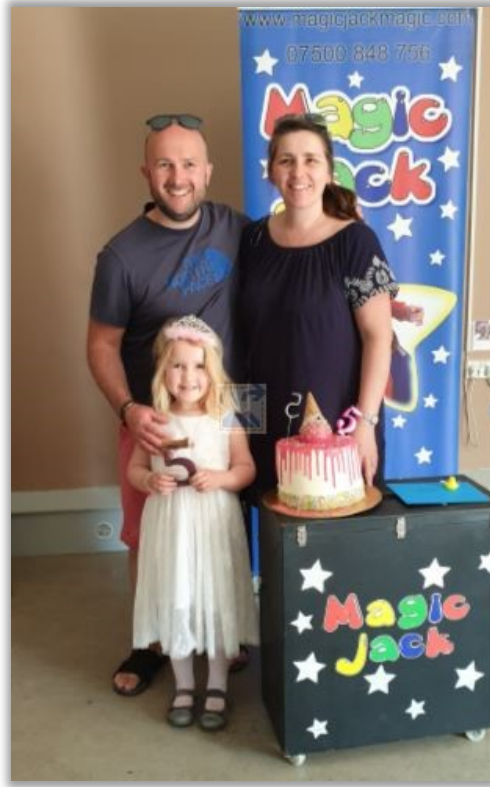
We wanted to make sure that everyone would hear about Fabry disease. We wanted to make sure that not only doctors, not only government, not only patients, but the entire society would understand how crucial that process was for patient's lives and health. Within less than a year Fabry disease was the main topic of medical conferences, conversations on the radio and television, publications in the press. Only in 5 months, there were more than 600 publications in the local media. We were almost everywhere. Everywhere Fabry disease was highlighted and discussed as the biggest priority for the near future. The campaign that we initiated, called "Fabry – a burning problem" became a great success. Photographs of Fabry patients and their families were taken by great polish photographer – Jacek Poremba, and published as 6 moving stories of the pain, burning and suffer. All of them were printed as standard postcards that could be sent to anyone – journalists, physicians, politicians. All of them were also available in a bigger format as an exhibition during medical conferences, debates or even on one of the Warsaw streets .



We definitely made a lot of noise across the country. At the same time, a number of meetings between our Association and Ministry of Health was organised. The noise in media was not enough and we wanted to be an equal partner for discussion in these processes. We also sent hundreds of letters to polish politicians asking them for any kind of support. To be honest, that was the most intense, difficult and busy time in my life. And not only mine, since a lot of patients got involved in these activities. Actually, without teamwork, probably most of it would never be possible. The more we tried, the bigger our hope was. And clearly, all of this effort paid off when in August this year the official announcement was stated – Treatment of Fabry disease in Poland is finally reimbursed. Although, it is indeed a great success for our association, I think we don't see it this way. I must admit, I felt huge relief when I heard this information for the first time. Then, there was pure joy to let everyone know that we got this. And after that, I realised that there is still a lot of work to do. However, we got the crucial starting point to make lives of all Fabry patients in Poland better and better. We now have the option for treatment. And most of all we can be sure that after last year, rare diseases are not so mysterious anymore. We can believe that it is going to be only better and better. Finally, we know that Fabry disease in Poland is like thousands other diseases - it can be treated.

*Ana Moskal, President of the Association of Families with Fabry Disease in Poland*

## Fabry Awareness Month 2019 - Photo Contest Winner



I was diagnosed back in 2011, a week before I started IVF. I really nearly pulled out of the procedure as I was so scared about the diagnosis and what it might mean for any should we be so lucky to have children.

Goodness me, after a whole lot of heart searching and deciding there are some illnesses that can never be picked up, had I been able to fall naturally I never would have terminated so I went ahead. Three cycles later, many losses along the way, our little Blossom is now 5.

She's put the sunshine back in our tummies, she is a constant delight and whatever the future holds, as true as the sun will rise, and fall every day she brings optimism and happiness into our little corner of the world.

The photo is me, my husband and Blossom at her magical birthday party last week. Every day with people who love you for being you, look out for you and you love back makes you the richest person in the world. I am content and happy with what I have. I try not to worry about the future, it's still 50:50 whether B has inherited the faulty gene from me. I think she has as she sometimes says her hands and feet are hot and itchy. We've been advised to not test her until she's older unless of course the symptoms take over.

My Fabry did not affect my medical when we were going through the adoption process before B arrived. I'm hopeful that one day soon we will be able to start the adoption journey again and we will be a family of four.

We are a vegetarian household and I'm sure my diet over the past 24 years will have helped my lysosomal storage disease. I'm not defined by Fabry, I'm defined as a wife and a mummy to Blossom, who is my end and my beginning.

That's how I start my every day.

Thanks for reading

Georgina Cartwright

Southampton UK

***On behalf of the entire FIN board, we would like to thank you and congratulate you Georgina, for sharing your story and sending in this beautiful picture of your family! We wish you and your family all the best for the future!***

## Summary Reports workshops Fabry Expert Meeting 2019

This year's Fabry Expert Meeting also had a new initiative – a day dedicated to workshops. We had topics such as “Understanding the psychological needs of Fabry disease”, “Symptom management”, “Let's talk Fabry men” and “Let's talk Fabry women”. All the patients and their caregivers were divided in to smaller groups to discuss one particular topic and each participant could choose three workshops to participate in. We have summarized the outcome of the workshops to share this with you. Rest assured we have taken note of your feedback and will be organizing workshops again at the Expert Meeting in Amsterdam next year with similar and new topics!



### “Let's talk Fabry Men”

*Moderators: Martynas Davidonis and Jack Johnson*

As board members of the Fabry International Network (FIN) Martynas Davidonis and Jack Johnson were very eager to facilitate the Let's talk Fabry Men workshops and go over anything that is related to being a Fabry male. This is a topic we have wanted to address with the members of FIN for a long time and during the last Fabry Expert meeting we finally got the chance to do so. Of course we must understand Fabry men not only from the perspective as a man who has Fabry disease, but also from the perspective of a man without Fabry who has met a woman with Fabry disease. It is important to recognize it does not really matter at what level she may be affected because they could have children one day that are affected differently and this would impact both of them.

***“A man must have a boy, plant a tree and build a house”.***

We prepared a simple so called agenda for our workshop and it was based on a proverb which is common in many countries: “A man must have a boy, plant a tree and build a house”. This very briefly explains pretty much everything required to be a man: to be strong mentally and physically, be able to make decisions, enjoy successes and cope with the unhappy things as well as be able to enjoy life with a woman keeping in mind all these important aspects.

Despite our agenda we didn't strictly stick to it and discussed many additional topics. The discussions were dynamic, unique to each group and frequently full of positive emotion. These very fluid conversations reinforced the importance of open and frank discussions because we have many similar experiences providing a strong foundation for understanding, but we also bring our own distinctive points of view that allows others the ability to see things in a new and enriching light. By sharing both our similar and diverse experiences we are able to forge greater understanding between patients and caregivers as well as build ourselves and our community into stronger survivors.

# FIN Workshop: Symptom Management/sharing experiences

*Moderators: Mary Pavlou and Lut De Baere*

Lut and Mary led the Symptom Management/sharing experiences workshop in 4 different groups of 12 – 15 people each. At the beginning of each workshop we made sure confidentiality rules were well understood by every one and we agreed to share only the overall outcome of the workshops and no names or personal stories.

Every one shared their personal story and even though every story was unique there were also similarities. We talked about the age of diagnosis which for the majority was at the age of puberty and most commonly after the diagnosis of a sibling or parent. The latest diagnosis was at the age of 24 years old.



The experiences when growing up were common for all. The majority experienced problems with activities and sports in general and discomfort while exercising outdoors during warm weather. Pain, GI problems and hypohidrosis were all common symptoms. In some cases parents were advocates for their children during school time to ask for permission to go to the restroom or to be excused from gym and sports. Additionally alterations in diet were made by the majority to avoid GI problems. The most common was to avoid foods that you know you had problems with, eat gluten and lactose free foods, keeping a food and symptom diary etc. An interesting one was to eliminate all food and slowly start adding a new group of food (like fruits, proteins, dairy products, etc) every other week.

In order to deal with pain, other than painkillers or tegretol which is commonly used by Fabry patients most patients are doing acupuncture, TENS machine, dancing, ice patches, yoga or even CBD oil. Some using an app to monitor the pain in between doctor's visits. Weather always makes pain and discomfort more difficult as many report "always being freezing". Hot weather is also a problem. For migraine and headaches using a TENS machine, meditation, diet alteration and medicines seem to help.

For insomnia, tinnitus and anxiety that some reported, having melatonin pills (insomnia) and practising yoga helps a lot to assist walking or even steroids injections in the ear for tinnitus. Many experience hearing loss and for that reason they are using hearing aids.

High blood pressure, cardiac and respiratory problems are most likely to be treated with pharmaceutical medicines and machines (pacemaker). Some reported to have more Fabry Patients with a kind of asthma or cough in their countries.

Fatigue is another common symptom among Fabry patients that affects work, and decreases motivation for sports and exercise. Some are working part time, work from home, need more rest days than others or need more days to recover and/or take B12/magnesium pills.

Mental health is a big issue because everybody needs to be "normal" but the body doesn't allow it. Also some felt guilty because the diagnosis was only made after the elder sibling was already affected a lot by Fabry.

As for monitoring and doctors visits some can talk to their physician every 2 weeks before, during or after ERT. They all reported being well monitored but due to lack of time by the doctor in depth discussion of the symptoms do not take place. In some countries the visit is once or twice a year undergoing annual tests (ECG, hearing tests, eyes, kidneys, heart etc). Home treatment is available in many countries. In Taiwan a heart biopsy is an obligation for non-classic Fabry patients in order to receive ERT. Group therapy with psychologist is not common but it helps.



It's not new that not many doctors are aware of FD. There are not so many experts in the field and that's the main reason for delay in diagnosis or misdiagnosis as a psychological problem.

Overall the workshop was a great experience that everybody enjoyed very much and wants repeat next year.

# “Understanding the psychological needs of Fabry disease”

*Moderators: Nadia Ali, Ph.D. Health Psychologist, Emory University, Atlanta, GA, USA and Anna Meriluoto*

The FIN annual meeting in Barcelona this May was amazing! Many thanks to the Board for inviting me and so many thanks to everyone who participated in the Members Workshop “Understanding the psychological needs of Fabry disease.”

Twenty-five members participated in the workshop, in three separate groups led by myself and FIN Board member, Anna Meriluoto. We started each group by going over the rules of confidentiality (i.e. no one may share another person’s story outside of the group). Then members introduced themselves and shared a fun fact with everyone.

Next, each group came up with a list of psychosocial issues relevant to living with Fabry disease. We also contributed as a group to a list of coping skills for dealing with life with Fabry disease!

Anna and I promised that we would share the combined lists from all the groups with everyone in the next FIN newsletter, so here we go! (Of note, while not everyone with Fabry experiences ALL of these issues, someone with Fabry has experienced each one of them and we respect them.)

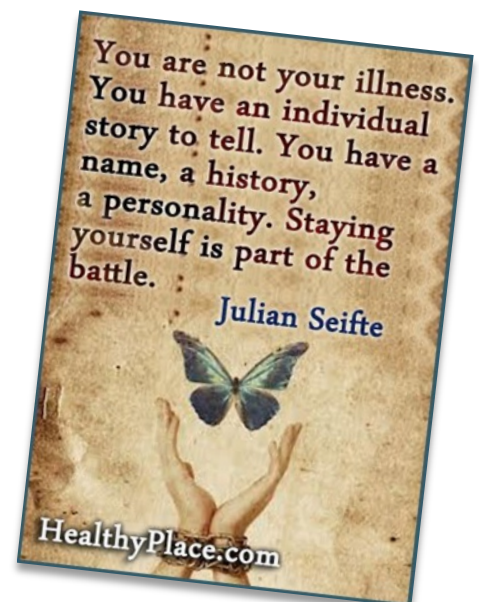
## Psychosocial issues relevant to living with Fabry

- Desire to be understood
- Guilt at passing Fabry on to your children
- How to tell family (extended family)
- Shock at learning diagnosis
- Uncertainty regarding disease progression
- Shame at being sick
- Being labelled a victim
- Career complexities
- Injustice (Is it fair that I’m sick and everyone else is healthy?!)
- Dating (When do you tell someone you have Fabry?)
- Deciding whether or not to have children
- How to parent children equally (between those who have Fabry and those who don’t)
- Teenage rebellion & transitions
- Denial (unable to accept limitations, overworking ourselves)
- Closed in your own thoughts / Over-thinking
- Invisible disease (you don’t look sick, so people don’t believe you have a chronic disease)
- Depression
- Anxiety & Fear
- Being blamed by other people (financial cost, societal cost)
- Loneliness, Isolation
- Desperate, feeling like giving up

## Coping Skills for dealing with life with Fabry

- Building community and connection with other people with Fabry
- Hope for the future / Faith
- Advocacy work
- Compassion / Relative adversity (Empathy for others and the idea that there’s always someone worse-off than you are)
- Reframing your identity (from that of a sick person to a strong person who happens to have Fabry)
- Finding your own passion (and engaging in it!)
- Humor / Laughter
- Therapy in all its many forms (Counseling, Art therapy, music therapy, pet therapy, nature therapy, sports therapy)
- Mindfulness and meditation (Clearing your brain)
- Compartmentalizing Your Pity (give yourself permission to feel it intensely for certain amounts of time and then put it away again)
- Learning to say ‘no’
- Napping
- Quality vacation time away from daily life
- Knowledge is Power
- Invisible disease (don’t have to tell people you have Fabry unless you want to tell them)

We sincerely hope you’ll find the list helpful and encourage you to try out different coping skills to find the ones that work for you!



# "Let's talk Fabry Women"

*Moderators: Anne Grimsbo & Erica van de Mheen*

Anne and Erica have put together a list of topics that were discussed during each of the workshops

## **Workshop/Group 1**

- Thorough personal introduction of the participants
- Different solutions in different countries. Even changes from region to region
- Descriptions of different ways of living with Fabry
- Personal reflections about being a patient, being a mother for children with Fabry
- Holiday and treatment. Your treatment should fit your life – your life should not fit treatment
- Relations to your family, caregivers, etc
- Corporation with your spouse to keep the family going

## **Workshop/Group 2**

- Introductions
- Being a mother of children with Fabry
- Options in prenatal diagnosis
- Women's right to treatment
- Difference between classical Fabry and non-classical
- Trouble with authorizations
- Organizational challenges
- Information leaflets and material
- Visible disease versus non-visible disease
- Women symptoms in particular
- Skin challenges

## **Workshop/Group 3**

- Introductions
- Family pattern
- Challenges at work
- GP's vary in knowledge in Fabry
- Differences between regions in countries.
- Home infusions.
- Criteria for treatment.
- Diversity of disease.





## **THE RARE FAIR 2019**

### **#WelcomeToTheTable**

FIN has been very fortunate to have been able to join the 2019 Rare Fair which took place on Sept 27th. Topics on community building, drug development and clinical trials, advocacy and legislation were on the agenda. Excellent speakers, great occasion to network and interesting materials to read.

The Rare Fair is the only 100% virtual, 100% inclusive event for the rare disease community. When you join The Rare Fair, you can log on from anywhere in the world to enjoy unprecedented access to this global event.

Their virtual event operates exactly like a traditional health fair with keynote speakers in a dedicated theatre, breakout rooms, an exhibitor hall, networking rooms, gamification, and patient education resources.

At The Rare Fair, patients and caregivers can network directly with physicians, medical providers, non-profit organizations, and other rare stakeholders. Our goal is to bring together the best rare resources available so attendees can add those resources to their own diagnosis management network.

This year's theme was #WelcomeToTheTable, illustrating their belief that every rare patient, caregiver, and advocate deserves a seat at the table and a voice in the conversation.

All those handouts and information packs you typically have to drag around events? You can keep them all organized in the Digital Binder. After the event ends, you'll have 30 days to log back on and access content you saved, view content you missed, or connect with exhibitors you want to follow up with.

[Visit the website](#)

## FOCUS ON FABRY LEADERS AROUND THE WORLD

### Interview with Mikhail Kuznetsov from Road To Life Russia



**When was your national patient association established?**

In December 2012.

**What was the reason for establishing a patient organisation?**

Establishing a patient organization for Fabry disease and other rare disease patients “Дорога к жизни” (“Road to life”) was related to the fact that my son has Fabry disease.

**What are the vision and mission of your association?**

Support, consultations, legal help regarding access to treatment for those affected by rare (orphan) diseases.

**How many members are there?**

There are 15 members (patients and their family members).

**What do you consider to be the major achievements or activities you are proud of?**

Rare disease day, meetings for patients, doctors and government officials, yearly rare disease congress.

**Can you name some challenges that your association is currently facing?**

That some government officials do not understand that rare diseases are very important problem and it has to be solved right now.

**Can you name some future goals or plans?**

Yearly rare disease congress “Дорога к жизни” (“Road to life”), round tables, trainings for patients, education for patients and their families.

**Has your association had issues with the national health system or insurance problems?**

There were problems back in 2010 when medications for Fabry disease in Russia was not present in the list of 24 rare diseases. This issue was solved after year 2011 and now every year the access to treatment becomes better.



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## FOCUS ON FABRY LEADERS AROUND THE WORLD

### Interview with Mikhail Kuznetsov from Road To Life Russia



**How would you describe the current treatment situation in your country?**

*I would evaluate treatment for Fabry disease 5/5.*

**What are the major issues with Fabry disease in your country?**

*There is a problem with getting disability for Fabry patients assessed when they still have good health condition until the certain age. And if the patient does not get the treatment on time there might be a need of dialysis or organ transplant and only then there is a possibility to get the disability assessed.*

**How would you further raise global awareness for Fabry disease?**

*I would suggest demonstrating video clips on TV, organize informational flash-mobs about the Fabry disease, to create various informational shields.*

**What are your suggestions for future projects for FIN?**

*To organize an Expert meeting in Russia.*

**What would you like to explain to other people / doctors / nurses / decisionmakers about Fabry disease?**

*It is very important to diagnose and start treatment for Fabry disease as early as possible because if the patient starts treatment too late there will be improvement but the organ transplant might be necessary to make life better.*

**What kind of information are we still missing about Fabry disease?**

*Will the cure for Fabry disease be possible in the near future?*

**What is your most effective means of taking your mind off Fabry disease?**

*Not to think about the disease but to live active and healthy life.*

**Is there anything else you would like to add?**

*I wish prosperity and fruitful work for the FIN team, expansion of possibilities and connecting Fabry patient organizations worldwide.*

**Thank you for your time Mikhail!**



**Mikhail hands the pen to Najya Bedreddin from APMF in France**

## *New publication*

### Symptoms and Quality of Life in Patients with Fabry Disease: Results from an International Patient Survey

Olivier Morand . Jack Johnson . Jerry Walter . Leone Atkinson . Gregory Kline . Aline Frey .  
Juan Politei . Raphael Schiffmann

#### Abstract

Introduction: Fabry disease is an X-linked lysosomal storage disorder caused by a deficiency of  $\alpha$ -galactosidase A. Symptoms include neuropathic pain and gastrointestinal problems, such as diarrhoea. To inform and support the design of a Phase III clinical trial for a new oral treatment for Fabry disease, this study evaluated patients' experiences of Fabry disease symptoms, the impact of symptoms on their quality of life, and their views on participating in clinical trials.

[Click here](#) for the full article

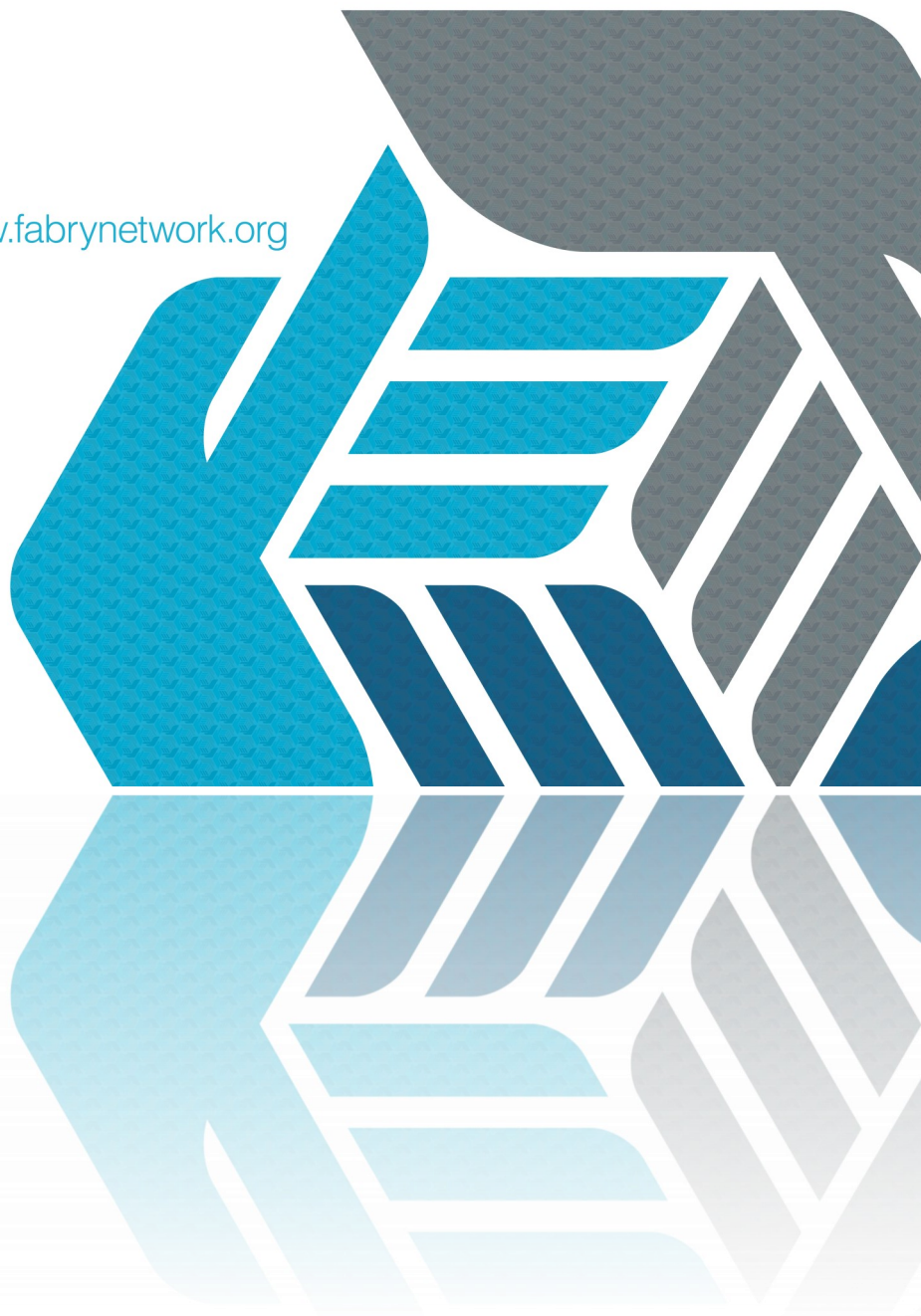




**FABRY**  
International Network

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[www.fabrynetwork.org](http://www.fabrynetwork.org)



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# Newsletter



## December 2019

### Contents

- Expert Meeting 2020
- Thank you Fabry Korea
- Takeda Charitable Access Program
- Clinical Trials Page—MyTomorrows
- Announcement vacancy FIN Board
- Fabry Findings Issue 2
- “I Hand the Pen To” - Interview with Fabry Leaders around the world
- Clinical trials Worldwide - myTomorrows
- New publications

### A word from the President:

*Dear All*



*The last Newsletter of 2019!*

*In this edition we continue to share a lot of information and announce a vacancy for the FIN Board. We would also like to draw your attention to our next FIN Expert meeting, next year in Amsterdam. We look forward to seeing you all at the meeting.*

*FIN recently added a page dedicated to clinical trials worldwide on the website. Make sure you don't miss it! Of course, you will find a lot of other news which I am sure you will find an interesting read.*

*On behalf of the FIN Board I would like to thank everyone for all their support and interaction, and for the information you have provided us to share in our 2019 newsletters. The Fabry International Network Board wishes you a very enjoyable festive season.*

*In order to make this newsletter as informative as possible we are always welcoming news and information from you as (family of) patients, patient organizations and patient representatives. Please send it to our mailbox: [info@fabrynetwork.org](mailto:info@fabrynetwork.org)*

*Please enjoy reading the FIN newsletter!*

Lut, FIN president



## **FIN**

# **EXPERT MEETING 2020**

### **SAVE THE DATE**

**We look forward to welcoming you in Amsterdam to  
the FIN Fabry Expert Meeting on April 24th-26th, 2020!**

**The meeting will take place at the [Steigenberger Hotel](#) near Schiphol Amsterdam**

### **Preliminary program**

**Thursday April 23rd: arrivals**

**Friday April 24th: workshops and AGM followed by dinner**

**Saturday April 25th: Expert Meeting followed by dinner**

**Sunday April 26th: Expert Meeting ending at 3pm**

### **Confirmed speakers**

Prof Germain

Dr Metha

Dr Ali

Dr Körver

Dr Böttcher

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**[Please find here the link to the booking form](#)**



FIN was invited to attend the first Fabry Korea Patient Meeting hosted in Seoul, Korea. It was our pleasure to present the Fabry International Network to them. We would like to thank the Songsuk Welfare Foundation and Fabry Korea for being such a wonderful host and all of the members for welcoming us to the meeting.



감사합니다.



Dear Readers

Fabry International Network is happy to remind you that the Takeda Charitable Access Program is open and accepting applications.

Deadline for applications is January the 6th of 2020 therefore we encourage you to contact your healthcare specialists in the eligible countries and ask them to complete the online application form for the patients they know.

**Important: the application form should be completed only by a treating doctor of patients suffering from Fabry, Gaucher or Hunter's (MPS II) disease.**

If the patient meets the treatment eligibility requirements, please see the list below to determine eligibility by country and condition:

- Egypt (Fabry and MPS II)
- Pakistan (Fabry, MPS II, Gaucher)
- Morocco (Fabry)
- Tunisia (Fabry, MPS II, Gaucher)
- Palestine (Fabry, MPS II, Gaucher)
- Tanzania (Fabry, MPS II, Gaucher)
- Botswana (Fabry, MPS II, Gaucher)
- Ethiopia (Fabry, MPS II, Gaucher)
- Sudan (Fabry, MPS II, Gaucher)
- Ghana (Fabry, MPS II, Gaucher)
- Nigeria (Fabry, MPS II, Gaucher)
- Kenya (Fabry, MPS II, Gaucher)
- Indonesia (Fabry, Gaucher)

Physicians may find eligibility details and access the application form on the following website: [https://webportalapp.com/sp/takeda\\_cap\\_application](https://webportalapp.com/sp/takeda_cap_application)

For those not familiar with the Takeda Charitable Access Program, it is designed to fulfill Takeda's long standing commitment to help patients live better lives. The program does this by providing the opportunity for otherwise underserved individuals to access life-saving therapies.

Should you have any questions please contact: [martynas@fabrynetwork.org](mailto:martynas@fabrynetwork.org).

## **FABRY INTERNATIONAL NETWORK BOARD MEMBER**

### **NOMINATION PROCESS**

There is currently one vacancy at the FIN Board. Nominations for a new Board Member are now open and an election process is herewith given together with the nomination form. If you wish to put your name forward for the selection process to become a FIN Board Member, could you please complete the nomination form, remembering that you need one supporter. Please return your completed form to, FIN **by March 1st 2020.**

[www.fabrynetwork.org](http://www.fabrynetwork.org)

#### **Nomination Criteria**

Nominations will be considered from people who meet the following criteria:

- Have an active interest in the Fabry International Network and are prepared to invest the necessary time and effort to fulfil the duties attached to the appointment.
- They belong to a Fabry Patient Organisation.
- They are supported by at least one colleague.
- Comfortable in the use of the English language.

#### **Nomination and selection procedure**

- The nomination and selection procedure leading up to the election at the AGM is set out below:
- Nominations must be returned to the FIN Office by March 1st 2020.
- Nominations must be supported by at least one colleague.
- Nominees will attend phone or in person interviews with members of the current FIN Board
- A selection of suited candidates will be presented at the annual AGM where the new board member will be appointed by the AGM attendees.
- All nominees will be advised of the outcome.

#### **Role of the Board Member**

- The role of the Board Member is to support the entire organisation through the process of governance.
- The Board plans FIN's future mission and priorities, it monitors performance and measures outcomes.
- The Board is responsible for all strategic planning and monitoring tasks and actions undertaken.
- Board members are individually responsible for participating in online and F2F Board Meeting. The language of these meetings is English and the appropriate expenses will be reimbursed (F2F).

**[Click here for the nomination form](#)**

## FABRY FINDINGS

Scientific publications that are translated into lay language made available to the Fabry Community;  
A new article will be announced in the FIN newsletter.

In this issue, we explore pregnancy and Fabry disease, and present the latest findings on the use of ERT during pregnancy

This issue is based on the publication:

***Enzyme Replacement Therapy During Pregnancy in Fabry Patients : Review of Published Cases of Live Births and a New Case of a Severely Affected Female with Fabry Disease and Pre-eclampsia Complicating Pregnancy.***

**Madsen CV1, Christensen EI2, Nielsen R2, Mogensen H3, Rasmussen ÅK1, Feldt-Rasmussen U4.  
Published in JIMD Reports**

### Fabry Findings

Issue No. 2 | Winter 2019



Current guidance issued by the regulatory authorities advise to use treatment for Fabry with caution<sup>1,2</sup> or to avoid its use altogether<sup>3</sup> during pregnancy.

These recommendations have been based on the limited amount of data available for these treatments in pregnant women or from the findings of earlier studies conducted in animals.

There are a number of published reports describing cases where treatment with enzyme replacement therapy (ERT) has continued during pregnancy.

In this issue, we explore pregnancy and Fabry disease, and present the latest findings on the use of ERT during pregnancy

#### What is Fabry?

Fabry disease is a rare, genetic condition which is estimated to affect around 1 in 100,000 people.<sup>4</sup>

In Fabry, an absence or reduced level of an enzyme called  $\alpha$ -galactosidase A ( $\alpha$ -Gal A), means that the body cannot break down certain types of fats, called globotriaosylceramide (GL-3 or Gb3) and plasma globotriaosylsphingosine (lyso-Gb3), and Gb3 builds up in a variety of cells in the body.<sup>4</sup>

This build-up causes damage to tissues and organs and leads to a range of symptoms and complications, which vary from one person to another.<sup>4</sup>

#### Fabry in women

Until 2001, women were considered, by medical professionals, to be 'asymptomatic carriers' of Fabry disease: that they can pass the disease on to their children without experiencing any of the symptoms themselves.<sup>5</sup>

It is now widely recognised that most women with the mutation in  $\alpha$ -Gal A (**heterozygous disease**) experience the disease in the same way that men with the mutation do; with a significant burden of disease and impaired quality of life.<sup>6</sup>

**A woman has heterozygous disease**

She has the Fabry gene on at least one of her two X chromosomes (XX), one inherited from her mother, one inherited from her father

 Information on how Fabry is inherited can be found on page 2.

 Information on the variability of Fabry symptoms and complications, and what effect pregnancy has on these, can be found on page 3.

[Click here](#) to read the second issue of Fabry Findings

The Fabry Findings articles are published on the FIN website

<http://www.fabrynetwork.org/fabry-findings/>

We encourage you to translate  
the articles into your own language so these can be shared in your community.

## FOCUS ON FABRY LEADERS AROUND THE WORLD

### Interview with Najya Bedreddin from APMF



#### When was your national patient association established?

*I have been a member of the association since its creation in 2005. In 2015, I joined the office as vice-president and I was appointed president in 2016.*

#### What was the reason for establishing a patient organisation?

*Having Fabry's disease, I was happy to find an association solely dedicated to my illness. After 10 years, I proposed to get involved in the association to make up for all that the association had brought me in support.*

#### What are the vision and mission of your association?

*The mission of the association is based on the following eight pillars:*

- *Inform members of the Association and patients with Fabry Disease about advances in knowledge of this disease*
- *Provide answers to questions asked by patients or those around them, validated by a scientific committee*
- *Create a friendly bond between the members of the association*
- *Listen to patients with Fabry disease and their entourage*
- *Promote research*
- *Act with public authorities*
- *Promote knowledge of Fabry disease*
- *Cooperate with other associations*

#### How many members are there?

*There are 255 members.*

#### What do you consider to be the major achievements or activities you are proud of?

*The association has organized an annual patient meeting every year for the past 14 years. Patients meet for a weekend annually in a city in France. This meeting provides information on the disease as multiple physicians are participating. It is also an opportunity to meet other people who live with the same illness and therefore feel less alone. This meeting is a real success because on average 120 people are present. And an journal is published in January which provides information on the latest advances in Fabry disease and which sheds light on the day to day organisation of our patient association..*



#### Can you name some challenges that your association is currently facing?

*The challenge of the association is to find the necessary funding to support the association and to organize the patient meeting weekend.*

#### Can you name some future goals or plans?

*Our goal is to work on the care pathway in Fabry disease. We are thinking about a project to improve the care pathway in Fabry disease.*

#### Has your association had issues with the national health system or insurance problems?

*Unfortunately when a person has Fabry disease it's very difficult - if not impossible - to get insurance to get a home loan. Resulting into patients not being able to buy a property because their credit scoring is refused.*

...



## FOCUS ON FABRY LEADERS AROUND THE WORLD

### Interview with Najya Bedreddin from APMF

#### How would you describe the current treatment situation in your country?

*All three treatments are available and reimbursed in France. There is a charge of approximately 50 euros per patient annually.*

#### What are the major issues with Fabry disease in your country?

*In France, the management of the disease when it is detected, is well supported because there is a reference centre for Fabry disease in Paris and various hospitals throughout France that take care of patients. The difficulty remains to diagnose the disease because with the different symptoms it is necessary that a physician thinks of this disease and researches it. Some patients go to several doctors and the disease will be found after many years because no one will have thought of looking for Fabry.*

#### How would you further raise global awareness for Fabry disease?

*By training physicians.*

#### What are your suggestions for future projects for FIN?

*A meeting with associations so that associations can exchange the different documents and tools they have created for their members.*

#### What would you like to explain to other people / doctors / nurses / decisionmakers about Fabry disease?

*The pain is very important, as well as the fatigue which leads to a poor quality of life are not considered enough. Organ damage (heart, kidneys ...) can be measured but the quality of life is not taken enough into account.*

#### What kind of information are we still missing about Fabry disease?

*Will the cure for Fabry disease be possible in the near future?*

#### What is your most effective means of taking your mind off Fabry disease?

*Gene therapy is certainly the solution that will allow the disappearance of the disease.*

#### Is there anything else you would like to add?

*FIN is a great organization that allows international exchange which is essential in rare diseases. Thank you all for your efforts!*

**Thank you for your time Najya!**



**Najya Bedreddin hands the pen to Stefania Tobaldini from AIAF ONLUS**



FIN recently started a collaboration with myTomorrows. myTomorrows helps patients with Fabry disease navigate the world of pre-approval medicines. The myTomorrows platform offers information and facilitates access to Clinical Trials and Expanded Access Programs available worldwide. They are dedicated to helping patients who have exhausted their regular treatment options.

myTomorrows' medical team is helping patients and their physicians. myTomorrows is an independent, globally operating organization based in the Netherlands. They provide patients with an unmet need, and their doctors, information about treatment options worldwide and facilitate access to medicines in development.

FIN and mytomorrows believe that everyone should have access to suitable treatment options.

Their goal is to ensure that patients and physicians don't miss out on treatment options due to lack of information, understanding and administrative hurdles.

[Click Here](#) to go the Clinical Trials Page on our website

Click on your country to get an overview of treatment options in development.



“Will you be attending the WORLD Conference in Orlando? We’ve heard from several patient organisations that they will be attending this year’s Lysosomal Disease Network annual WORLD conference in February. If you are attending, so are we! . For more information about this conference, please visit: <https://worldsymposia.org/>



Rare Disease day coming up on Feb 29th, 2020! Rare is many. Rare is over 300 million people around the globe. Rare is strong. The rare disease community joins together across borders and diseases to raise awareness and advocate for equity. Rare is proud. Show your support for the rare disease community with pride! The likely truth is that you know one of the 1 in 20 people affected by a rare disease. Raise awareness by showing your support on Rare Disease Day 2020!

*The next FIN Newsletter will be out at the end of March 2020. Please send your articles and contributions by email to Charlotte no later than March 15th, 2020. We look forward to publishing and sharing your latest news with our International Fabry Community!*

## **Gene Therapy/Editing Series 1: A Brief Introduction To Gene Therapy**

Bhavneesh Sharma - Registered investment advisor, biotech, contrarian

This is the first article in a series on gene therapy/editing which was first published on the Marketplace premium service last week. The next article in the premium service will discuss the various delivery systems for gene therapy/editing. After discussing the basics like delivery systems, manufacturing processes, etc., the series in the premium service will focus on landscape of gene therapy/editing in different genetic diseases. This will be followed by discussion of individual gene therapy/editing companies, their SWOT analysis, many of which already belong to the premium service portfolio.

[Click here](#) for the full article




## **Gene Therapy Series 2: Delivery Systems In Gene Therapy, Including Viral Vector**


Bhavneesh Sharma - Registered investment advisor, biotech, contrarian

Continuing with the series of articles on gene therapy, this article will discuss the various delivery methods for gene therapies with a focus on viral vectors. The article also discusses the classification of gene therapy methods by the site of delivery, and a comparison of the characteristics of viral vectors used in gene therapy. Examples of public gene therapy companies using different vector delivery systems for gene therapy are also provided.

[Click here](#) for the full article



*All of us send you warm wishes for a happy, peaceful holiday season and a prosperous new year. From us to you...may your holiday season be filled with peace, happiness and hope. Thank you so much for the gift of our community.*



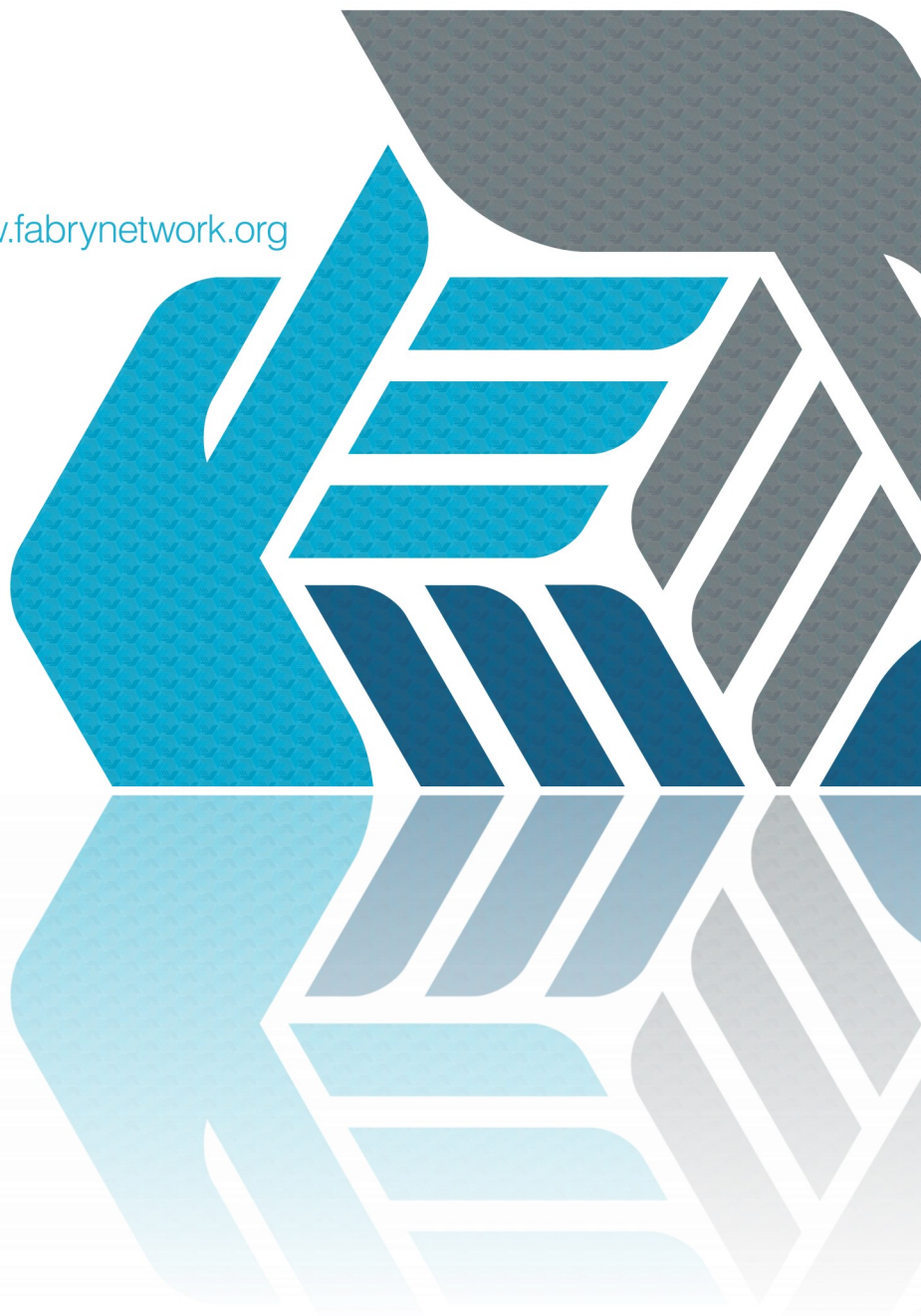
*May the joy of the season stay with you throughout the year.  
Best wishes for the holidays and the coming year from the entire FIN Board.*



**FABRY**  
International Network

FACILITATE | SUPPORT | ENABLE

[www.fabrynetwork.org](http://www.fabrynetwork.org)



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