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

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 the chance to 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
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
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.

Association of Families with Fabry Disease in Poland - Treatment of Fabry disease in Poland is finally reimbursed!
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
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 melatonine 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.
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.
- 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.
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
Symptoms and Quality of Life in Patients with Fabry Disease: Results from an International Patient Survey


Abstract

Introduction: Fabry disease is an X-linked lysosomal storage disorder caused by a deficiency of a-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