Newsletter

FABRY International Network

June 2020

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A word from the President:

Dear All

We proudly present the latest FIN newsletter.

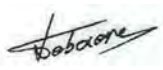
On June 10th, we held our annual general members meeting, although we would have loved to meet in person, we were very happy to meet many of you online.

In 2020 (and beyond) FIN will focus on projects and initiatives that bring patients together. Of course, this is part of FIN's mission. We feel now more than ever, there is a need for patients to be part of a wider community. We aim to do so through sharing best practices, facilitating connections between Fabry patients to help combat isolation and supporting local initiatives. In case you missed the AGM, we have included most of the information in this newsletter.

The new dates for the FIN Expert meeting in 2021 are now confirmed. Make sure you save the date!

In order to make this newsletter as informative as possible we always welcome news and information from you as a caregiver, patient, patient organization and patient representative. Please send it to info@fabrynetwork.org

Stay safe and healthy!



Lut, FIN president

THE JOURNEY OF LIVING WITH A RARE DISEASE IN 2030

Anna Meriluoto

In mid-May The European Organisation for Rare Diseases *Eurordis* organized its annual conference on rare diseases and orphan products. The conference was originally planned to take place in Stockholm, Sweden but due to the Covid-19 pandemic it was swiftly moved to be held completely online. It was a magnificent effort from Eurordis with more than 1500 participants from 57 countries attending the 2-day event and yet things were running very smoothly. The online platform was clear and simple to use and provided a lot of easy-access information and several interactive networking opportunities.

The ECRD is recognized globally as the largest, patient-led rare disease event in which collaborative dialogue, learning and conversation takes place, forming the groundwork to shape future rare disease policies. The backbone of the conference agenda was the Rare2030 –project led by Eurordis. Rare2030 is a foresight study that gathers the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations that will lead to improved policy and a better future for people living with a rare disease in Europe (www.rare2030.eu). It is of utmost importance for us all to identify the major trends and drivers that shape the future for rare diseases.

What are the major trends in the field of rare diseases in Europe then? First of all there is a rise of the pan-European multi-stakeholder networks to advance diagnostics, treatment and care. There is an unprecedented potential in the European reference networks that will definitely shape the future of rare diseases. The success will, however, depend on the activity of the member states as well as different stakeholders. The patients will get an earlier diagnosis, newborn, prenatal and pre-symptomatic, so the numbers of rare disease patients will grow. That will make the member states face new kinds of issues and problems, e.g. add strains on the health care budgets and the emergence of new care delivery models. New healthcare technologies will provide many beneficial developments such as telemedicine, virtual consultations and monitoring diseases with different health apps. However, access to new digital technologies will be slow and limited. They will also bring us new challenges and questions about data governance, ownership of our own data and what happens to it once it´s been fed into the new technologies.

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The future will bring us a change in the demographics of rare disease patients as all European countries are faced with a rapidly aging population. That will lead to greater co-morbidities and complexities including those for rare disease patients. This will strain the member states financially and we might see a greater variation in access to treatments and care resulting in more inequality for people with rare diseases. This might also lead to a system of prioritization. Keeping in mind that only 5% of rare diseases have an existing treatment today, we need to work towards creating a paradigm of inclusion, solidarity and fairness. We also need to make sure that the rare disease patients and patient groups are empowered and fully involved in all aspects of their disease management.

What should be the take home –messages from this conference? There are a lot of innovations in healthcare which promise positive develop for a diagnosis. Patients provide even more valuable input to all public sectors. Transformations in care for rare diseases provide better standards for medical and social care. Patients have an opportunity to receive better therapeutic development.

We are witnessing the beginning of the era of digital health technology to improve standards of care. And we have an action plan of accessible, affordable and available treatment of people with rare diseases by 2030.

As Benjamin Franklin pointed out: “If you fail to plan, you plan to fail.” I will finish with a quote from the Eurordis Chief Executive Officer Yann Le Cam: "We are preparing for the next decade. We are bringing forward solutions, created at the margins of society but becoming more mainstream. Rare2030 was the backbone of ECRD 2020. The outcome of the conference is a new impetus to initiate a new legislative framework for rare diseases**."**

The recording of all the conference sessions is available online until May 2021 for a small attendance fee. Find out more at https://www.rare-diseases.eu/ecrd2020-all-sessions-available-one-year/ If you are interested in viewing the conference posters, they are available at https://www.rare-diseases.eu/posters/

FIN EXPERT MEETING 2021

SAVE THE DATE

April 23rd - April 25th, 2021

Amsterdam, The Netherlands

The meeting will take place at the Steigenberger Hotel near Schiphol Amsterdam

ERT HOME INFUSION FROM MY PERSPECTIVE

Erica van de Mheen

Hi, I'm a 63-year-old Dutch woman diagnosed with classical Fabry disease. I have two daughters who are also Fabry affected and we live in the North of the Netherlands.

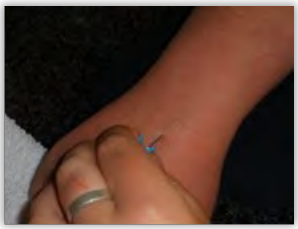
In January 2003 I started ERT to make sure that no more damage was done by Fabry disease. At the beginning I already made the decision to do it at home and do the infusion myself. I was so happy that this was possible in the Netherlands already then.

Frisian wooden shoes for a child, on my fence door

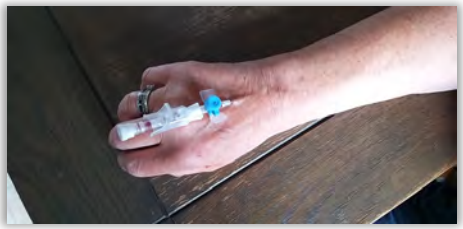
Before starting the home infusions the physicians had to be sure, of course, that there would be no infusion reactions and that you would be doing fine after the infusion so you always need to have your first infusions at the hospital. You also have to learn how to apply the infusion materials and to use the needle properly.

(Note: in the Netherlands it is possible to do the infusion completely by yourself, without any help from a trained nurse, but with help from a partner or a friend!)

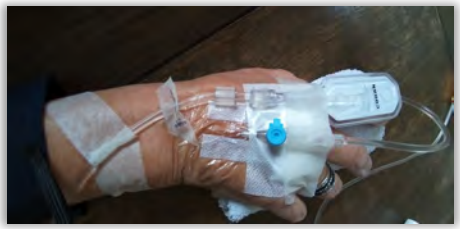
It is very important that your workplace is clean and safe. At my home, we do it at the dining table, with a nice white clean towel beneath. I prepare the infusion myself and I stick myself at the back of my hand with a small butterfly needle with help of a friend or my daughter to fix the IV. My youngest daughter started at sixteen and she also took her infusion at home, sometimes on the same day.



My daughter puncturing herself.



The nurse uses an IV-needle



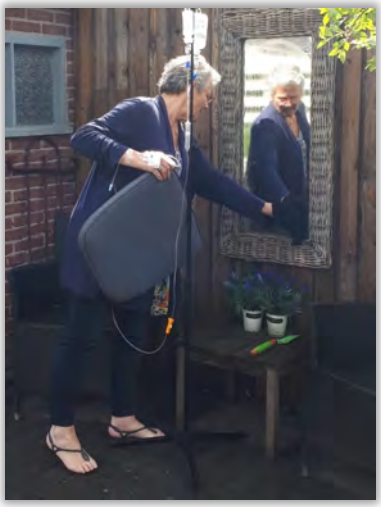
Completely fixed so I can use my hand

Since 2015 I live alone, my daughters (36 and 28 now) have their own houses and since then a nurse comes to my home to help me with the infusion.

I get all the required materials delivered at home. My medication for two infusions is also delivered once a month. The medication needs to be kept in the refrigerator to stay cool.

As a member of the Dutch Patient Organisation you receive a mobile IV-pole free of charge when you start ERT. FSIGN has done this since 2003 and it is much appreciated, because insurance does not provide this needed instrument for free. You can take this with you everywhere, even camping or on a boat trip. I like to sit in the garden or go to my sisters' or my daughters' home so they can assist me when the nurse is on vacation or otherwise not available. Yes…. all of this is possible over here.






It is my opinion that there are only **positive sides to home infusion**:

- You can do it at home in your own environment. You don´t have to go to a hospital, which might not be near your home. Some people have to drive far and for a long time.
- You can sit on your couch, watch a movie or your favourite series, sit at the table playing a board game with friends or family, sit in your garden when the weather is beautiful and so on.
- There is no need for a hospital bed, so you don't feel like a patient.
- You have no extra cost e.g. fuel and/or use of car, bus, train etc. and parking money.
- You can choose your own preferred time to infuse, even in the evening, during the weekend or some other time when the hospital is not open for day care....very important!
- Without the hospital visit the cost for home infusion versus day care is much lower.
  In The Netherlands, the treatment cost is approximately 8 times lower than getting treatment in a hospital, even with a nurse present at home.
- No need to worry about hospital bacteria.
- You don't have to miss work, school or other important things, which you might if you had to go to the hospital every two weeks!

As you can see.....I am a really big ***FAN*** of home infusion!

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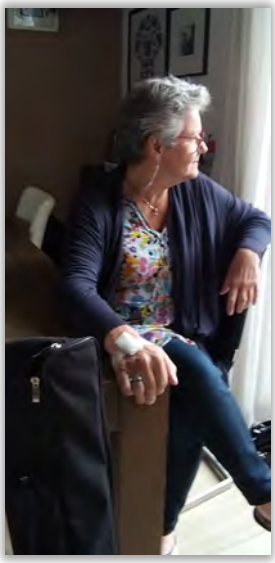
In my garden

On the sofa



All done

I would like to promote the option of home treatment for all Fabry patients worldwide and let everyone know how much it improves my quality of life. We are all aware that ***Quality of Life*** is so important for people living with a chronic disease.

We already have to sacrifice so much, lets make this one thing easier for all of us.

I encourage you to translate this report into your own language to share with your community as an example of what can be achieved.

Always willing to provide more info!

Yours sincerely

Erica van de Mheen
treasurer@fabrynetwork.org

FIN
AWARD

FIN wants to encourage the membership to organise new activities and initiate projects by contributing financially and giving a platform to share with the wider community.

FIN will award a patient (association) led initiative that informs and educates about Fabry and helps raise awareness by providing a financial grant*

The winning project will be announced at the Expert meeting in 2021

The activity or initiative should be:

- Educational and raising awareness
- Bringing patients together (face to face or virtually)
- Providing peer support

We encourage all out of the box ideas!

Download the application form here

**Terms & Conditions are stated in the application form*

COVID-19 and Fabry Disease

Since we wanted to keep our community informed during these uncertain times, we hosted our very first webinar in April, which was very successful. A second webinar took place in May. We were very happy to have our expert panel which consisted of Prof Germain, Prof Eyskens, Dr Ali and Dr Ortiz presenting valuable information to our members.

The recordings are available on our YouTube channel.

Dr. Nadia Ali, Ph.D Emory University, USA

Click here to listen to the presentation

Prof. dr. Francois Eyskens UZA, Belgium

Click here to listen to the presentation

Prof. Alberto Ortiz MD, Ph.D, Health Research Institute of the Jiménez Díaz Foundation Madrid, Spain

Click here to listen to the presentation

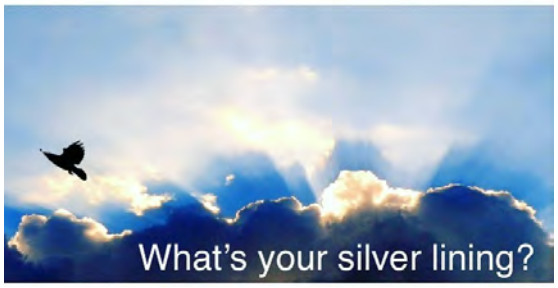
Prof. Dominique P. Germain, MD Ph.D National Center for Fabry disease, France

Click here to listen to the Q&A

Over the course of 2020 we will organise additional webinars with topics from the expert meeting program in close collaboration with Fabry experts. We will also focus on topics where we see patients as more than just Fabry.

COVID-19 and Fabry Disease

Many of our members organised webinars of their own to inform the community along with additional heart-warming initiatives .



Every cloud has a silver lining means that even the worst events or situations have some positive aspect. Sometimes it is hard to find the positive but it is at these times when it is most important to do so.

We were forced inside to look inward... we asked our community to send in their silver linings through COVID-19 and you can feel a shared sense of togetherness, strength, and resilience.

From making noise for front line workers, reinventing creativity, and taking time to slow down and be present. Adversity shows us what we're capable of and magnifies our strengths. Watch the video here to bring some inspiration to your day.—*Julia Alton*

A card with a heartwarming message

For the Dutch patient association, one of our members put together a special card, that we sent out to all our members as we were not able to meet face-to-face.

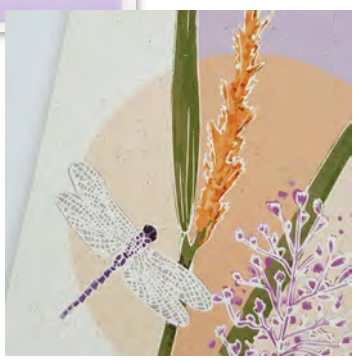
It is a design from her own collection 'Plukgeluk' (Choose luck)
The colours are completely adapted in the colours of our own corporate identity.

The design on the back is completely handwritten and designed by Francine, just like the personal postage stamp, with a small piece of art on the address label, which is reflected on every lavender coloured envelope. This completes the image and creates a unique experience.

A special project for people with unusual challenges to encourage them. Printed on paper with a subtle fibre, stylish and environmentally friendly from Dutch soil, sent in an environmentally friendly envelope.

The Dutch text translates into English:
Wishing you lots of strength during this difficult time, we are thinking of you.

Given the responses, the gesture was much appreciated.
Small gesture, big impact - *Erica van de Mheen*



The COVID-19 health emergency was a very complex moment for all of us. Many of us had to spend a long time in solitary confinement without having contact with other people. Medical checks in hospitals had also been canceled. For this reason AIAF has created the "AIAF IN CONNECTION" project: a virtual space with professional support. It has been an opportunity for sharing and talking with patients and families involved in Anderson-Fabry disease to feel less alone and stay in touch with each other. Since April we have organized five webmeetings with patients.

We worked with a psychologist, a lawyer and the AIAF Scientific Committee in order to discuss patients' needs related to the COVID-19 emergency, provided information about the absence from work due to COVID-19 risks and updated them on the scientific news related to Fabry disease and answer their questions. Thanks to this project AIAF has been in contact with about 40 Italian families during the COVID-19 pandemic.

During the COVID-19 pandemic, AIAF also made recommendations for the therapy management available to patients in collaboration with the scientific committee.

The Italian version is available here

Stefania Tobaldini, President of AIAF Onlus

fabrysuisse

Fabry Suisse organised a webinar for their members as they could not meet in person.

Mrs. Faiza Kaddour-Gysi Specialist Psychologist for Psychotherapy FSP, Solievo.net - interdisciplinary centre for mental health, spoke about "Accepting the disease and dealing better with the disease". PD Dr. Albina Nowak, Senior Physician, Endocrinology Clinic Rare diseases, University Hospital Zurich presented "New horizons in therapy for Fabry disease" - *Beate Krenn*

MPS LISOSOMALES *New logo!*

On May 4, we started a bi-weekly cycle of conferences, in collaboration with Sanofi Genzyme, a cycle of virtual conferences on Lysosomal Storage Disorders. Webinars are open and free to all those interested and it can be followed online from any device and from any country.

The contents of each session will be led by different experts in these pathologies and will be focused on offering theoretical and practical content, always oriented to support the patients and families during the COVID-19 pandemic.

The topics discussed are of general interest related to lysosomal diseases as well as specific topics related to Fabry disease. After each conference there is time allowed for attendees to ask questions to the experts.

These sessions took place in May and the beginning of June

Session 1: May 18 "*Our strengths facing adversity", Eduardo* Brignani, Professor in psychology. Reflections to help us get in touch with our capacity to start again, even in a complex and uncertain reality, to live each day as one new opportunity.

Session 2: June 4. *"Monitoring of lysosomal diseases and treatments in the Covid-19 situation",* Dra. Mireia de Toro, neuropaediatrician at the Vall d'Hebrón Hospital, Barcelona. *"Home therapy"* Javier Salguero, director of the Ashfield Healthcare Patient Unit.

Session 3—: June 17: labour related topics. Lorenzo Pérez, Lawyer specialized in labour issues.
The topics of the following sessions are to be determined. Suggestions made, are being evaluated.

Session 4 will be on 23th of July, 7PM CET, the importance of the family tree in Fabry disease will be presented by Dr. Roberto Barriales from the Inherited Cardiovascular Disease Unit, A Coruña Hospital.

Check the website www.mpsesp.org for the recordings of the sessions

We encourage all of you to participate — *Jordi Cruz*

In Belgium, care packages were sent to all of the members.

The packages consisted of a mouth mask and a small bottle of hand sanitizer. Our organization contacted all of the members directly by phone. We wanted to make sure everyone of our members were safe and felt supported during this difficult time. - *Lut De Baere*

BOKS VZW

Thank you for all your submissions for our
Let's get Fabry Creative contest!

We enjoyed viewing and reading your beautiful poems, videos, drawings, stories, paintings etc.

They truly were creative!

FABRY
International Network

Let's get Fabry creative

We are very happy to announce the winner of the contest:

Geonwoo Kim

Congratulations!

Geonwoo Kim wrote a beautiful poem.

FABRY AWARENESS MONTH
APRIL 2020

Winner Let’s get Fabry Creative Contest

봄

길고 긴 겨울 밤

잠 못 이루고 뒤척이다 깨어서

긴 밤 홀로 깨어있네

졸음에 겨워 설핏 잠들었다

이내 다시 깬다

졸다 깨다 반복하며 영영 오지 않을 것 같은 새벽을 기다린

다

길고 긴 겨울 지나

봄이 오고 쌓인 눈 녹듯

아픔도 봄 눈 녹 듯

스르르 녹아내리길...

Spring

In a long winter night

I´m awake after tossing and turning

I´m awake all night alone

Fell asleep for a while

Soon, I wake up again

I´m waiting for the dawn that never

seems to come, while dozing off

After passing through a long winter

Like spring comes and snow melts

Hope the pain goes away

Like snow melts in spring...

FOCUS ON FABRY LEADERS AROUND THE WORLD

Interview with

Mary Pavlou

The Panhellenic Association of Patients & Friends with Lysosomal Diseases "Solidarity"

Greece

When did you join your national patient association and what was the reason for joining?

I joined my national association in 2002 after I was diagnosed with Fabry disease. It was a difficult period for me and it was the only way to cope with the news of having a rare and serious disease. It was also a way to support my family as I learned more about Fabry through the association. Since then I'm a board member and I actively support and help patients. I currently am the Vice President for our organisation, along with the rest of the board members we organize activities and projects to support, educate and empower our members. Through several activities we try to raise awareness for lysosomal diseases.

What is the vision and mission of your association?

The Pan Hellenic Association of patients and friends with lysosomal diseases "The Solidarity" was founded in 1997 in Athens, Greece and has 120 members. It is a non-profit organization acting as the sole representative of patients suffering from Lysosomal Diseases. Its main goal is to inform patients, relatives and carriers in Greece about diagnosis and the monitoring procedures of Lysosomal storage Disorders. The association was founded with the dream that all patients of lysosomal diseases would have access to treatment, early diagnosis and reimbursement of treatment from public insurance. A registry for rare disease patients was at the table early on. Moreover our continuous goal is to empower all our members.

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

Due to our Association's long and deep involvement and commitment to our patients, we are recognized in Greece as one of the leading associations concerning Rare Diseases and Lysosomal one's in particular. We are also founding members of the first official EGA, now IGA, and a proud member of Fabry, Pompe and MPS International. My colleagues are very proud of having a representative in FIN.

We are co-founders of the Greek Alliance of Rare Diseases and a permanent member of EURORDIS by participating annually in their meetings. We are actively and publicly speaking about the rights of patients with rare diseases. We organize press conferences, meetings both with doctors, nurses and patients and we publish articles in newspapers about our problems. We try to engage our members to learn their disease, speak up to their doctors about their problems and to reach out to us should they face any problems.

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FOCUS ON FABRY LEADERS AROUND THE WORLD

Interview with

Mary Pavlou

The Panhellenic Association of Patients & Friends with Lysosomal Diseases "Solidarity"

Greece

Last year to celebrate International Fabry, Pompe and MPS Day we attended an inclusive art theatre play where the actors were people with disabilities. The play was simultaneously explained for people with difficulties in both hearing and vision. In the past and for several years we provide psychological support for our members through a help line, organise face to face sessions for young adults and parents. We try to be present at all the major conferences to learn more about the diseases we are representing and travel all across Greece to support and get to know our members better.

Last but not least, the participation of our Chairman as the key speaker and representative of all suffering from Rare Diseases in a debate in 2017 in the Greek Parliament concerning the Greek centers of expertise of Rare Diseases and also a debate about the new established offices concerning the rights of patients in hospitals is another achievement we are proud of.

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

One of the biggest challenges that we are facing is that we are not getting any younger! We try hard to engage young patients and involve them more in activities we organize. We hope someday to step down and let them lead the association. It's always refreshing to have the next generation around. Our doctors are getting older too. This is why we believe that the centres of expertise can play an important role to continue development in care, early diagnosis, state of the art monitoring and the support in psychological and social level. On an operational aspect it is always difficult to work with limited budgets. Last but not least our deepest concern is to raise awareness for Lysosomal Storage Diseases from the general and scientific public as Lysosomal Disorders still remain in obscurity and need to be promoted in every possible way.

Can you name some future goals or plans?

We want to continue our current projects and add some more! The pandemic has changed our plans but we still hope we will organize yet another meeting to empower our members. We would also like to start to provide social and psychological support to them again.

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

Currently two ERT's are reimbursed and one oral treatment is at the stage of becoming reimbursed in Greece. Our patients are treated in general hospitals and we don't have the option of home treatment. Moreover a Fabry centre of expertise needs to be established soon.

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

Time of diagnosis is still an issue. Most of our patients went through several doctors before they received their diagnosis. The lack of a Fabry expertise centre as well as the lack of an institutional framework for home treatment, I think, is an important issue we need to address soon .

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FOCUS ON FABRY LEADERS AROUND THE WORLD

Interview with

Mary Pavlou

The Panhellenic Association of Patients & Friends with Lysosomal Diseases "Solidarity" Greece

How would you further raise global awareness for Fabry disease?

That's an ongoing project we are all working hard to achieve. I believe that patient advocates and representatives should have a larger presence at scientific congresses. Rare diseases such as Fabry disease will never be well known if we don't speak about them publicly. Fabry should be diagnosed as early as possible . We have to educate the general public that there are diseases like Fabry that are not obvious at first sight. Meaning that we suffer from all kind of symptoms but because someone else is not able to see them that doesn't mean they are not there. Using social media platforms could be a useful tool to do so.

What are your suggestions for future projects for FIN?

As I'm a member of the FIN board and I know some of our exiting new projects I think it's better to wait and see!! With this opportunity I would like to say that working for FIN it's a once in a lifetime experience. We are a very strong team, we have passion for what we do and I'm so proud to be a part of it.

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

I reckon that we still need to learn more about the social and psychological burden Fabry patients are facing. Those are aspects that we haven't addressed in the past. In addition the role that food can have in gastrointestinal problems and in general how the dietary habits can affect symptoms. I would also like to see more publications on female life events, and the transition from childhood to adulthood. I think it's a highly underestimated problem and needs to be addressed.

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

Reading is my most beloved thing to do to get my mind of Fabry. Talking to friends, swimming and a trip to the countryside is always a good idea!

Is there anything else you would like to add?

Don't be afraid to raise your voice for your rights. We all have the right to be living as equal members of the society with respect to our limitations. We all have dreams and we want them to come true.

Thank you for your time!

Mary Pavlou hands the pen to Ching-chang Hsu from the Taiwan Fabry Disease Patients Association

Anne Grimsbo

Martynas Davidonis

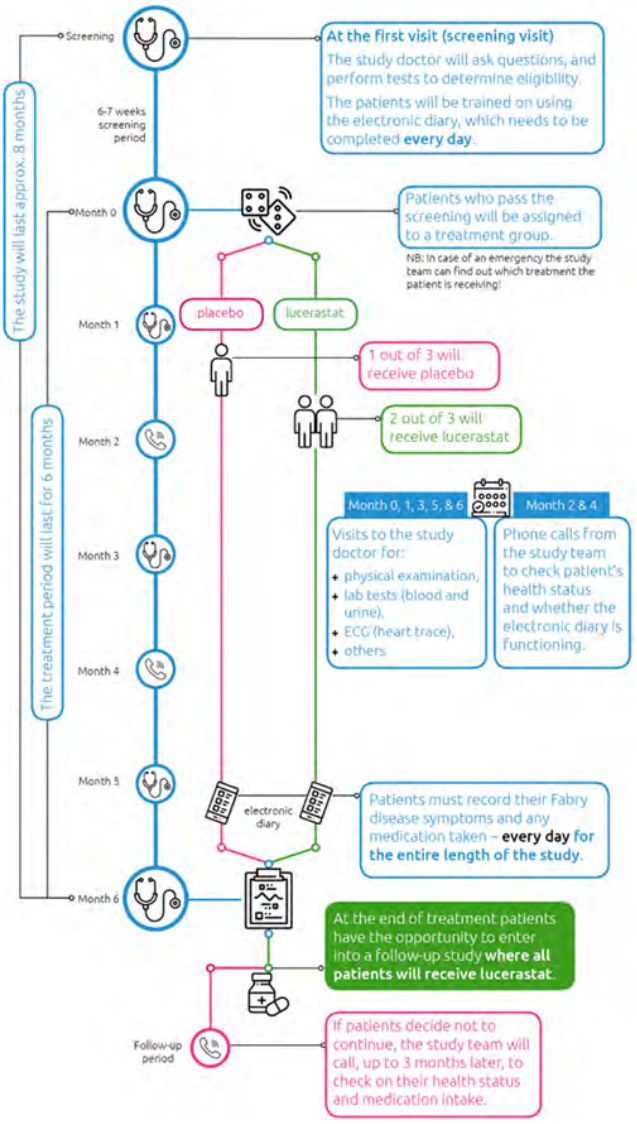
FIN regrets to inform you that there are 2 board members resigning; Anne Grimsbo and Martynas Davidonis.
We would like to thank Anne and Martynas for their efforts and valuable contributions to our community. We wish them all the best.

We have sincerely enjoyed the time that we have spent working with you. Thank you for all the guidance, support, and encouragement that you have shown over the years.

MODIFY - A phase 3 study investigating a potential new oral treatment for Fabry Dis-

The ongoing MODIFY study is investigating the effectiveness and safety of lucerastat, a potential new oral therapy for Fabry disease. MODIFY is currently enrolling participants at medical centers in North America (USA & Canada), Europe (UK, Belgium, Netherlands, Poland, Austria & Germany) and Australia. During the COVID-19 outbreaks in these countries, enrollment of new study participants was put on hold to enable medical staff to focus on the treatment of patients with COVID-19. However, individual medical centers are now progressively lifting these restrictions and enrollment into the MODIFY study is recommencing. In addition, participant enrolment will soon be initiated at new sites in five additional countries across Europe (Ireland, Italy, Spain, Norway & Switzerland).

What is neuropathic pain?
a type of pain which feels like burning, shocks or shooting, stabbing, tingling, and/or pins and needles in the hands and feet.
...sound familiar?

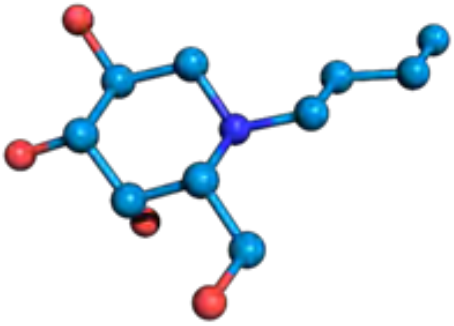


Lucerastat is administered in the form of oral capsules taken twice a day. Any adult with Fabry disease, irrespective of the type of genetic mutation that they have, may be eligible to participate in the MODIFY study. The key entry criterion is neuropathic pain, defined as sensations of burning, shocks or shooting pain, tingling, pins and needles, stabbing, and/or numbness in the hands and feet. Neuropathic pain may be permanent or occur randomly, and may be triggered by heat or cold, a fever, and/or physical activity.

MODIFY is a phase 3 study, meaning that lucerastat is now in the final stage of the clinical trial process that must be completed to evaluate the safety and efficacy of a new medicine before it is submitted to Health Authorities for review.

Participants in the MODIFY study have a 1 in 3 chance of being randomly assigned to receive placebo treatment. The capsules provided to participants assigned to receive placebo treatment will not contain any lucerastat. However, participants who complete the 6-month treatment period will have the option to enroll into an extension study investigating the long-term effects of lucerastat treatment. In the extension study, all participants will receive lucerastat. Treatment in the extension study can be continued for up to 2 years.

MODIFY - A phase 3 study investigating a potential new oral treatment for Fabry Disease



Graphical representation of the lucerastat molecule

The MODIFY study is sponsored by Idorsia Pharmaceuticals Ltd., a pharmaceutical company based in Switzerland (www.idorsia.com). Idorsia will organize and pay for travel, including air fares and hotel accommodation if required, even if study participants live far away from the nearest participating medical center in their country. For more information about the MODIFY study, visit www.modifyfabry.com, where patients with Fabry disease can submit a request to be contacted by the medical staff of one of the medical centers conducting the study in their country.

Additional information about MODIFY, including a complete list of participating medical centers in each country, can be found on the U.S. government`s ClinicalTrials.gov registry (use this link www.clinicaltrials.gov/ct2/show/NCT03425539 or search using the identifier number NCT03425539).

Two news releases issued by AVROBIO one of which reported on Fabry and cystinosis data, and the other on preclinical Pompe disease data.

Avrobio would like to take this opportunity to summarize these updates. We recognize that this is a lot of information. We will be hosting a call in June to review these updates, as well as answer any questions our communities may have. It's important to note that AVROBIO therapies are investigational, and their safety and efficacy are still being evaluated in clinical trials.

AVR-RD-01: AVROBIO's investigational gene therapy for Fabry disease

- Nine patients with Fabry disease (four patients in a Phase 2 trial and five patients in an investigator-led Phase 1 trial) have been dosed in two ongoing clinical studies of AVR-RD-01. The Phase 2 trial continues to enrol participants; the Phase 1 trial is fully enrolled.

- This news release includes interim data on the four patients dosed in the Phase 2 trial of AVR-RD -01.

- In particular, the interim data show sustained alpha-galactosidase (AGA) enzyme activity and consistent trends in other measures up to 22 months after treatment for the first patient dosed.`

- Additionally, the first patient with Fabry disease treated with platoTM gene therapy platform show a reduction in globotriaosylceramide (Gb3) one month after treatment, as well as, significantly higher white blood cell counts and AGA enzyme activity three months after treatment compared with the same timepoint for the other patients in the Phase 2 trial who were treated with the academic gene therapy platform.

- There have been no safety events attributed to AVR-RD-01 drug product in either the Phase 1 or Phase 2 trial as of the safety cut-off date (Nov. 29, 2019).

Chiesi Global Rare Diseases, in collaboration with Protalix BioTherapeutics, have submitted a Biologics License Application (BLA) to the US FDA for pegunigalsidase alfa for the treatment of Fabry Disease.

CARMIEL, Israel, May 28, 2020 /PRNewswire/ -- Protalix BioTherapeutics, Inc. (NYSE American: PLX) (TASE: PLX), a biopharmaceutical company focused on the development, production and commercialization of recombinant therapeutic proteins produced by its proprietary ProCellEx® plant cell-based protein expression system, or the Company, together with its development and commercialization partner Chiesi Global Rare Diseases, a unit of Chiesi, an international research-focused healthcare group, today announced the submission on May 27, 2020 of a Biologics License Application (BLA) to the U.S. Food and Drug Administration (FDA) for pegunigalsidase alfa for the proposed treatment of adult patients with Fabry disease via the FDA's Accelerated Approval pathway. Pegunigalsidase alfa, or PRX-102, was granted Fast Track designation by the FDA in January 2018. Pegunigalsidase alfa is the Company's purposefully-designed, long-acting recombinant, PEGylated, cross-linked α-galactosidase-A investigational product candidate.

The BLA submission includes a comprehensive set of preclinical, clinical and manufacturing data compiled from the Company's completed Phase I/II clinical trial of pegunigalsidase alfa, including the related extension study succeeding the Phase I/II clinical trial, interim clinical data from the Phase III BRIDGE switch-over study and safety data from the Company's on-going clinical studies of PRX□102. Upon the BLA approval, if approved, the Company will be eligible to receive a milestone payment from Chiesi.

"We are grateful for the assistance the FDA provided leading up to the submission of this BLA via the Accelerated Approval pathway, and we look forward, together with Chiesi, to working with the FDA as we seek marketing approval for PRX□102," said Dror Bashan, Protalix's President and Chief Executive Officer. "Together with Chiesi, we thank the investigators and study participants who have made reaching this milestone possible and have supported Protalix in our commitment to bringing this new treatment option to the Fabry patient community."

"The submission of this BLA to the FDA represents a significant milestone for our Global Rare Diseases division that was established earlier this year to strengthen Chiesi's focus on making a difference for patients living with rare diseases around the world," said Giacomo Chiesi, head of Chiesi Global Rare Diseases. "Our partnership and active collaboration with Protalix are a great example showing how we can leverage Chiesi's global reach and decades of experience in drug development to support patients and their families living with Fabry disease and many other devastating rare diseases."

Read the full press release here

This is an important step toward providing treatment options to patients.

Our thanks go to all those who are participating in the clinical trials – they are the ones who made this possible and deserve ours and the community's thanks.

FABRY INTERNATIONAL NETWORK BOARD MEMBER

NOMINATION PROCESS

There are currently two vacancies at the FIN Board. Nominations for a new Board Members 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.

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.
- Belong to a Fabry Patient Organisation Board or organization administrator.
- Have the support of at least one colleague.
- Are 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.
- 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 Awareness Month at Sangamo THERAPEUTICS

The next FIN Newsletter will go out end of September. Please send your articles and contributions by email to coordinator@fabrynetwork.org no later than September 15th, 2020. We look forward to publishing and sharing your latest news with our International Fabry Community!

FACILITATE | SUPPORT | ENABLE

FABRY
International Network

www.fabrynetwork.org

Fabry International Network

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Charity Registration Number 04080030 The Chamber of Commerce Drenthe The Netherlands