5th FIN Expert Meeting
19 & 20 May 2017
At the Royal Olympic Hotel
Athens Greece

www.royalolympic.com
email info@fabrynetwork.org to reserve your place
A Word from the President of FIN

This has been a most productive year for FIN with so many of its members working energetically in their respective countries. We appreciate this is not easy, particularly when the Fabry Patient Organisation relies on volunteers who are Fabry patients, family and supporters. It was wonderful to see so many FIN members at the Expert meeting hosted in Tokyo, Japan in May and the FIN Board look forward to giving you a very warm welcome at the FIN Expert Meeting in Athens in May 2017. To help with the organisation we encourage you to plan early to participate in this meeting dedicated to living with Fabry disease.

FIN applied to Genzyme's PALS Award programme earlier in the year and we are eagerly awaiting to hear whether we have been successful. The grant application is to provide a two day fully funded Fabry orientated educational workshop for the leaders of ten global Fabry Patient Organisations. If we are successful we plan to hold this workshop immediately prior to the Fabry Expert Meeting in May 2017 in Athens so if you are interested in participating in the Workshop you may wish not to book your flights until after we hear the outcome of the PALS Award application in January 2017.

I also want to use this Fin e-News to tell you that after a robust selection procedure Martynas Davidonis from Lithuania was appointed to the FIN Board. Welcome Martynas.

Christine Lavery MBE

Novel Treatment for Fabry Disease:

IV administration of plant derived alpha-gal-a enzyme safety and efficacy, 1 year experience by Prof R Schiffmann, USA

Fabry Disease (FD) is an X-linked disorder. PRX-102, a novel PEGylated enzyme for the therapy of FD expressed in a plant production system, is comprised of 2 submits covalently-linked via a 2-kDa PEG moiety resulting in a stable homo-dimer. This interim report is a 1 year clinical experience summarising _ 15 patients years treatment.

I will not give the technical details of this, but just a free report. PK results indicated a plasma half-life of approximately 70h and a substantially higher AUC. A total reduction in Gb3 inclusion in PTCs of 75,5% and 86,5% was achieved by 0,2 and 1 mg/kg, respectively, in kidney biopsies (BLISS). AI doses presented reduction in biomarkers, Gb3 and Lyso Gb3, stability in annualized eGFR and in cardiac MRI, and an improvement in MSSI. PRX-102 was well tolerated, with a 95% incidence of mild and moderate, adverse events. Only one of the patients experienced hypersensitivity. 19% developed treatment induced antibodies.

PRX-102 has unique PK properties which result in available enzyme throughout 2-wek infusion intervals that offer optimal opportunity for attenuating disease progression. The majority of adverse events were mild and moderate with limited formation of antibodies in long term data. In addition, to the various efficacy parameters indicate a potential positive treatment effect.

Lut De Baere

Migalastat for Treating Fabry disease

The Fabry international Network is pleased to announce that NICE have supported the recommendation for Migalastat to be used as a treatment option for Fabry disease in patients over the age of 16 years living in England. NHS England now have 3 months to comply with NICE’s recommendations and to ensure access to treatment is available for those individuals who meet the clinical criteria for this technology.

To read the full document, please click the link below.

Migalastat is an oral therapy and could be assessed as a treatment option under clinical guidelines for Fabry patients over the age of 16 years with an amenable mutation. If you would like more information about this therapy, please discuss with your specialist Fabry consultant.
AVROBIO, a clinical-stage biotechnical company developing transformative cell and gene therapies targeting cancer and rare diseases, today announced its launch plans. The company’s priority is to accelerate development of two novel cell and gene therapies pioneered within the labs of Dr Christopher Paige and Dr Jeffery Medin (now at the medical college of Wisconsin) at the University Health Network (UHN) in Toronto, ON.

Phase 1 programmes will be in the clinic by early to mid-2016 in both acute myeloid leukaemia (AML) and Fabry disease. The company will simultaneously work to expand its proprietary cell and gene therapy platform to treat additional indications.

Cell and gene therapies represent a new paradigm in human health, with the potential to deliver dramatic disease-modifying effects with long lasting, durable impact. Underlying these advances are a deeper understanding of cell biology, immunology and a newer generation of vector designs enabling safe and effective delivery of therapeutic genes targeted to specific cells.

AVR-02 is designed to deliver lasting benefits for Fabry disease patients. The company’s approach is to genetically modify a patient's own cells by adding a functional copy of the faulty gene. CD34+ hematopoietic stem cells are genetically modified to express the enzyme alpha-galactosidase A. The modified cells are then infused back into the patient via a one-time procedure.

The objective is to deliver long lasting or permanent, continuous elevation of endogenous enzyme thereby significantly improving patient outcomes and eliminating onerous lifetime biweekly intra-venous infusions of enzyme replacement therapy.

US Olympian, and rare disease patient, Cody Miller won a bronze medal at the 2016 Paralympics in the Men’s 100 meter breaststroke. With a time of 58.87 seconds, Cody finished just behind Adam Peaty of Great Britain and Cameron van der Burgh of South Africa. That was the only individual race Cody is scheduled to be in but look for him to compete later in the 400m relay. Congratulations, Cody.

Miller was diagnosed with Pectus excavatum at a young age. The hallmark of the condition is a sunken appearance of the chest. The condition also limits the person’s lung capacity forcing them to take more breaths than an average person. Though not all bad, Cody Miller actually has his diagnosis to thank for starting his career in swimming. At age 8 he started swimming because of his condition to monitor his heart and breathing- now he is a 2016 Olympic swimmer.

The shocking part is he is not just a swimmer, but a breaststroker. The world's best breaststrokers are 6’3” to 6’5”, and he is 5’10”. Yet Miller takes longer and fewer strokes than anyone. Miller never let his condition define who he was or stop him from going the distance. He became a junior national champion, set national age group records (15-16) in the 100- and 200-yard breaststrokes and was chosen for the national junior team. 2016 will be his first Olympic appearance. In tests at the U.S. Olympic Training Centre in Colorado Springs, Colorado, he was the most symmetrical swimmer that the judges have ever seen. Possibly due to his caved-in chest.

**Pectus excavatum**, also known as sunken or funnel chest, is a congenital chest wall deformity in which several ribs and the sternum grow abnormally, producing a concave, or caved-in, appearance in the anterior chest wall with an incidence of 1 in every 400 to 1000 live births. It causes respiratory issues and deficient lung capacity. Severe cases of pectus excavatum can eventually interfere with the function of the heart and lungs. The cause of pectus excavatum is not known.
Sunday September 4th I'm waiting for the departure of my flight to Rome. In the meantime, I have the opportunity to look at the program of the SSIEM congress, the World Congress for everyone who professionally deals with metabolic diseases. Tomorrow is actually a day of pre-meetings, which is very interesting for BOKS (Belgian patient organization for metabolic diseases). The program includes sessions from E Hod, MetabERN and E Imd. About MetabERN, I will explain in more detail below. The next few days are fully planned with sessions on various metabolic disorders. There are many parallel sessions, so choices must be made. It's actually easy, because although there are many abstracts in the accompanying book, there is only one session specifically devoted to Fabry Disease, under the "New therapies for lysosomal diseases". I will therefore display these below. This is an extremely tough congress, not only because of the many very interesting presentations, but also for the side activities, like personal meetings with doctors, researchers and companies. Very tiring but worth itiqué.

MetabERN
As mentioned in the previous FIN Newsletter, Prof. Scarpa is the coordinator of MetabERN. I am responsible for the patient representatives. On Monday, September 5th MetabERN was presented very extensively to the audience. The following day there were two shorter sessions following in the official program of SSIEM. Prof. Scarpa gave a presentation on what MetabERN is; a project of the European Commission, how it is organized, where we are now and where we work towards. The most important thing we need to remember here is that MetabERN is patient centered.

I have known Prof. Scarpa for a very long time. During, or perhaps it was in between, one of the EU workshops to establish the ERNs, Prof Scarpa asked me if I wanted to join the MetabERN and represent the patients in it. Later Eurordis made a call for volunteers to work within the different ERNs, in which I was elected as one of the 6 metabolic epag members.

As foreseen, in the MetabERN patient organisations and patient representatives, play an important role. The Patients Board will work closely together with the medical Executive Board. And patient representatives will be engaged at each level within the structure of the MetabERN.

Of course the main ultimate goal consists of having patient representatives fully involved so that they can act as a communication and information source from professionals to patients and vice versa. We already made a database of the European and international metabolic patient organisations. After sending out an information letter to all known European metabolic patient organisations, we made a list of candidates who are willing to be involved/engaged in this Network. Here are some numbers so it becomes a little bit more concrete for you. At the moment, we have identified 7 international and 4 European patient organisations and we have 6 epag members. We hope that all these 17 groups will be very active so we can achieve a lot together.

For several diseases or disease groups we already have patient organisations who are willing to share their experience with us and provide us more detail about the specific needs of their group. In total we have for the 7 subgroups 30 engaged European patient representatives over 12 European countries and 8 non-European followers.

Looking ahead there is a huge task waiting for us, like preparing a survey so we can identify the needs of the patients for each disease, but also the differences in patients' needs in the different European countries.

To conclude I'm happy and thankful that we, as patient representatives, have a great and unique opportunity to make a real difference in improving the quality of life for metabolic patients. We as patients are looking forward to the further development of MetabERN.

Lut De Baere
"On the 23rd to the 24th of September the Danish Fabry Patient Organization had their yearly outing at Brogarden in Funen. 21 members out of 55 took part in this event.

Friday night we had a lovely dinner and later a speaker, Freddy Meier who made a speech about how to keep joy, happiness and love as focus points in ones life.

Saturday morning we had patient debate groups. After that Anne showed pictures from the Fabry Expert Meeting in Japan. The event has had positive response from the participants. We look forward to next time."

The Greek Lysosomal Association, "Solidarity" is organising a one day scientific meeting which will be held at The Golden Age Hotel in Athens, Greece on 19th November 2016. Attending the meeting will be Greek doctors specialised in lysosomal diseases including Mr. Latrou MD, head of nephrology department, Fabry patients specialists, Mr. Anastasakis MD, cardiologist and a member of the Greek plan for rare diseases.

Also, we have the honour to participate as our gest speaker Mr. Cox MD, physician with great experience in lysosomal storage disorders and in Gaucher disease from UK. The meeting will attend patients from all over Greece.

If you have any news or events you would like to share in our next newsletter please send details to info@fabrynetwork.org

Fabry in Lithuania: the beginning

History of every disease always starts with the first patient diagnosed. For example Auguste Deter was the first patient diagnosed with Alzheimer's disease and Sir Augustus d'Esté was the first one known to have multiple sclerosis. I do not know exactly who was the first patient of Fabry disease in the world but I know for sure that in Lithuania the first patient was me. It was an exhausting, long and painful way to my diagnosis and treatment but despite all this I finally got what all other non-Fabry people have – the alpha-galactosidase enzyme in my bloodstream. This was a delightful event of my life....

But from the beginning:

I was born healthy and had a pretty much happy life for some time but then suddenly when I was four years old I began to get ill from time to time. I would have fevers but there was never any real explanation as to why. Occasionally I used to spend some time in hospital for treatment and examinations or because I was sick or depressed. When I was five or six years old during one visit to the hospital a young doctor asked me about the red dots I had on my chest. It appeared to me that neither the doctors, my mother or I knew what kind of rash we were dealing with. I don't remember the pain from these early days but I knew that it was present when I was twelve and later. As I grew older the pain grew with me... I took various drugs to cope with the pain but only later I found something that helped to relieve it. Of course during the adolescent period I was attending school except when I was seriously ill. From time to time I continued to spend weeks or months in hospital. I was of a small height also therefore I used to have somatotropin (a growth hormone) injections and this helped me to grow to 178 centimeters high. In 2003 I was hospitalized in an endocrinology department and it was at that time after hundreds of consultations I think I had my first consultation with a geneticist and one older doctor, when she saw me it was like she was hit by lightning and said..."It's Fabry". After that many interesting things happened in my life. I started medical school. The first year of that my health became really bad and creatinine of blood started increasing every visit to my doctor therefore I was put on hemodialysis in May 2006. Later that year I finally got a confirmed diagnosis of Fabry disease and after some time I had my first Fabrazyme infusion. When I started attending a medical unit for my infusions it there was no improvements for a while but everything changed when I had a kidney transplant in 2008 and simultaneously an enzyme effect appeared. Right now I work in the university with students and in June 2017 I will graduate from law school. This shows a great improvement of my health thanks to Fabrazyme and many good doctors. To be continued...

Written by the first diagnosed Fabry patient in Lithuania, Martynas.
WORLDSYMPOSIUM™ ANNOUNCES RECIPIENT FOR 2017 PATIENT ADVOCATE LEADER AWARD

WORLDSymposium will recognize Christine Lavery, MBE, Group Chief Executive for the UK Society for Mucopolysaccharide Diseases and President of FIN, with the 2017 Patient Advocate Leader (PAL) Award at the WORLD Symposium in San Diego 13-17 February 2017.

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.

Contact FIN for:
- Membership
- Latest News
- Information
- Connection
- Support

Board of Directors:
- Christine Lavery, President
- Jack Johnson, Vice President
- Lut De Baere, Director
- Anna Meriluoto, Director
- Anne Grimsbo, Director
- Mary Pavlou, Director
- Martynas Davidonis, Director

Main contact:
Toni Ellerton FIN Coordinator
Direct Line: +44 01494 54 67 60 Mobile: +44 (0) 7944 256672
Email: toni@fabrynetwork.org info@fabrynetwork.org

Postal Address:
Fabry International Network, MPS House, Repton Place, White Lion Road, Amersham, Buckinghamshire, HP7 9LP United Kingdom

www.fabrynetwork.org