A Word from the President

Dear Members of the Fabry International Network

Whilst this is the first FIN e-News of 2017 that doesn’t mean FIN has not been working hard on your behalf.

Since the turn of the year the FIN Board has met by teleconference with Shire Pharmaceuticals, Sanofi Genzyme and Amicus Therapeutics. At WORLD in San Diego in February FIN board members, Jack Johnson, Martynas Davidonis and I had the opportunity to meet with representatives from Genzyme, Shire, Amicus and Protalix to be updated on developments for Fabry disease in their companies.

With a new generation of Fabry treatments in the clinical trial pipeline from time to time there are opportunities for FIN to offer patient awareness of new developments. One such opportunity has come up in Europe and Fabry patient organisation representatives from Turkey, Germany, Czech Republic, Hungary, The Netherlands, Italy, Norway, Belgium and the UK have been invited to learn about one of a new ERT treatment which is currently under development for Fabry. Whilst we do recognise the demands on those leading Fabry patient organisations, many of whom combine this role with earning a living and family life it is disappointing to have so little response and find ourselves chasing.

With our FIN Expert Meeting in Athens only weeks away we are delighted that so many of our global FIN members will be joining us in Greece for what promises to be a thoroughly stimulating meeting and time networking. Toni Ellerton, FIN Co-ordinator is doing her best to make your time in Athens go as smoothly as possible but if you have any questions or concerns please do contact Toni on toni@fabrynetwork.org

Kind regards

Christine Lavery MBE

President

Contents

In this issue:

A Word from the President
AVROBIO raises $25m for clinical programs
Protalix shares additional positive results in PRX-102
Amicus files to market Japan
Japanese group reports successful repair of Fabry gene

NICE Backs Galafold in England
2017 FIN Expert Meeting in Athens, Greece
News from Fabry Patient Organisation’s
FSIG Expert Fabry Conference
First Scientific Program was held by the Greek Lysosomal Association ‘Solidarity’
AVROBIO, Inc., a clinical-stage biotechnology company developing transformative, life changing gene therapies for rare diseases and cancer, announced that it has raised $25 million in a Series A financing co-led by Atlas Venture, Clarus and SV Life Sciences. AVROBIO will use the proceeds from this financing to accelerate development of its clinical stage programs in Fabry disease and acute myeloid leukemia (AML), and to expand its pipeline in rare disease and solid and liquid cancers.

“AVROBIO’s highly innovative therapies offer potentially life-altering impact for patients following a single infusion of genetically-modified cells,” said Geoff MacKay, AVROBIO’s President and Chief Executive officer. “We are grateful for the funding and support we have received from our investors, as we continue to focus on displacing the standard care for patients with Fabry disease or AML through the development of these disruptive gene therapies.”

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. AVROBIO’s initial two programs are leveraging the established safety and effectiveness of ex-vivo gene therapy to provide Fabry and AML patients with new therapies that have the potential to significantly improve both their quality of life and lifespan.

AVROBIO’s phase 1 gene therapy to treat Fabry disease seeks to deliver lasting and meaningful benefits for Fabry patients. The patient’s stem cells are extracted and genetically modified by adding a new, functional copy of the faulty gene. The modified cells are then infused back into the patient via a one-time procedure. A durable elevation of endogenous enzyme is expected, with the potential to significantly improve patient outcomes and eliminate costly lifetime biweekly intravenous infusions of enzyme replacement therapy.

PROTALIX Shares Additional Positive Results in PRX-102


“We are pleased to announce additional positive results from our phase I/II Fabry clinical trial,” said Moshe Manor, Protalix’s President and Chief Executive Officer. “The efficacy results seen to date continue to be very strong with improvements or stabilization in efficacy demonstrated across all disease parameters giving us confidence in our phase III head-to-head study comparing PRX-102 versus Fabrazyme®. The safety data is remarkable with all adverse events being mild to moderate and transient by nature and with antibodies decreasing or being eliminated for the very few patients who tested positive. We believe the product profile for PRX-102 has the potential to be highly differentiated from currently available enzyme replacement therapies for Fabry disease and could impact significantly the lives of patients suffering from this rare, genetic disease.”

The phase I/II clinical trial is an open-label, dose-ranging study designed to treat up to 18 naïve male and female adult patients. The three dose cohorts include 0.2 mg/kg, 1mg/kg and 2mg/kg with intravenous infusions of PRX-102 every two weeks.
Amicus Therapeutics (Nasdaq: FOLD) plans to submit a Japanese New Drug Application (J-NDA) to request marketing authorisation for migalastat, an oral precision medicine for Fabry disease, in the first half of 2017.

Following a meeting with and written correspondence from the Pharmaceuticals and Medical Devices Agency (PMDA) in Japan, the J-NDA will be based on data from completed clinical studies with migalastat, including two pivotal Phase III studies, as well as a Phase I study that previously evaluated the pharmacokinetics (PK) of migalastat in Japanese volunteers. Taking into account data from Japanese patients included in the Phase III program and the similar PK properties in Japanese and non-Japanese individuals, the PMDA confirmed that these completed studies meet J-NDA submission requirements without the need to conduct an additional clinical study in Japan.

“The submission of our J-NDA will be an important milestone in our global strategy to pursue migalastat approvals for Fabry patients with amenable mutations as quickly as possible, in as many geographies as possible,” stated John F. Crowley, Chairman and CEO of Amicus Therapeutics, Inc. “With approximately 700 Fabry patients currently treated in Japan, we believe that a substantial number of Japanese Fabry patients could potentially benefit from migalastat”.

Japan represents the second largest Fabry market in the world by country. Amicus estimates that 35 to 50 percent of Fabry patients in Japan may have amenable genetic mutations.

As previously announced, the European Commission has granted full approval for migalastat, under the trade name Galafold, as a first line therapy for long-term treatment of adults and adolescents aged 16 years and older with a confirmed diagnosis of Fabry disease (alpha-galactosidase A deficiency) and who have an amenable mutation.

Amicus is currently also working with the US Food and Drug Administration (FDA) to determine the optimal approval pathway for migalastat for Fabry patients in the United States. An update is expected in the third quarter of this year.

Japanese Group Reports Successful Repair of Fabry Gene

At the Japanese Society for Genome Editing’s first conference, held in September 2016, in Hiroshima, focused on medical and healthcare applications, the group shared its plans to foster the development of appropriate research and applications, the group shared its plans to foster the development of appropriate research and application frameworks for human-use genome editing. This involves making changes to a person’s genetic code, which could potentially repair or enhance any human gene.

A team led by Professor Satoshi Gojo, of Kyoto Prefectural University of Medicine, reported that they have repaired the gene that causes Fabry disease through a technique known as CRISPR-Cas9.

Japan has a rule that bans genetic engineering to alter the human germline, which means making changes to human sperm, eggs or embryos, so these new methods will be discussed under ethical and regulatory issues by the Japanese Society for Genome Editing.
The National Institute for Health and Care Excellence for England is backing National Health Service use of Amicus Therapeutics' Galafold for treating the rare genetic disorder Fabry disease.

Fabry disease an inherited lysosomal storage disease caused by a non-functional or only partially functional enzyme called alpha-galactosidase A (alpha-gal A), which results in the build up of enzyme substrates that cause cellular damage in tissues throughout the body.

Galafold (migalastat) is an oral, small molecule drug designed to bind to the enzyme alpha-galactosidase A (alpha-gal A) as it is made, helping it to fold correctly and improving its function. It is a life-long treatment, that costs £210,000 per patient per year (excluding VAT and any discounts).

In a final evaluation determination, NICE says the drug can be funded on the NHS within its marketing authorisation, that is for use in people over 16 years of age with an amenable mutation, but only if it is provided with the discount agreed in the patient access scheme and only if enzyme replacement therapy (ERT) would otherwise be offered.

With the confidential PAS discount, Galafold has a lower total cost than ERT, and potentially provides greater health benefits than ERT, the Institute said, but also stressed that there remain limitations and uncertainties in the evidence presented for the drug.

Therefore, it is encouraging the company, NHS England and treatment centres to collect more evidence, particularly on the longer-term benefits of Galafold and ERT for treating Fabry disease, which should inform a future evaluation of the costs and benefits of all treatment options for the condition.
The Fabry Support & Information Group proudly announces the 2017 FSIG Expert Fabry Conference.

Friday April 28th—Sunday April 30th
Embassy Suites, Cincinnati - RiverCenter
10 East Rivercenter, Covington, KY 41011

This conference is especially for the US Fabry patient community with valuable, up-to-date information about Fabry disease. You will have the opportunity to network with others, learn from the experts, relax, and be a little pampered at the same time. There is an excellent line-up of informational presentations, breakout sessions, focus group discussions, awards, and surprises.


General agenda:
- Friday Evening—registration, Fabry 101 and welcoming social mixer
- Saturday Morning—registration and general session presentations
- Saturday Afternoon—concurrent breakout sessions
- Saturday Evening—dinner and awards
- Sunday Morning—special presentation on psychological impact and Fabry Disease

Conference hosted by the Fabry Support & Information Group
The first scientific program was held by the Greek lysosomal association “Solidarity” on Saturday 19th November 2016 in Athens, Greece. The room was full with patients and their families from around Greece who heard, many of them for the first time, speeches about their illnesses.

We had the privilege to receive the visit of Pr. Cox Tim FMedSci Professor of Medicine Emeritus Director of research Honorary Consultant Physician, University of Cambridge who talked about lysosomal disorders research and developing ERT for Gaucher and Pompe disease. His presentation was really impressive.

There were also specialists from Greece like biochemist Ms Mihelakaki who talked about laboratory diagnosis for lysosomal diseases and an exercise physiologist Mr. Terzis who present the benefits of exercise in Pompe disease.

The speech of Dr. Anastasakis PhD, scientific responsible for inherited cardiovascular diseases unit, about rare diseases in Greece and the European network for rare diseases was very interesting too. Also, Mr. Zafiriou, professor of pediatrics neurology, talked about treatment in pediatrics lysosomal diseases and Dr. Marinakis’, hematologist, talk about Gaucher disease and 20 years of treating Gaucher patients was really touching.

Furthermore Dr. Papadimas, neurologist, talked about MPS diseases and Dr. Panagiotou nephrologist, speech was about Fabry disease. In addition her work at local hospital in Thessaloniki, presented Ms. Drosou from a social’s worker point of view. A positive reception had the speech by Ms. Moustaka, psychologist who talked about psychology effects on patients with chronic diseases. Last but not least Chief Director on disability and occupational medicine, Ms. Niarchakou analyzed the current situation for people with disabilities in Greece and what they are entitled by the Greek government.

It was an opportunity for the Greek lysosomal community to hear from the experts about their diseases and how to handle their every day problems and to bring together specialist in the field of rare diseases.

The members of the Greek board were very satisfy with the participation and the acceptance that all the presentations had and they are looking forward for the next conference with more on lysosomal disorders.

Mary Pavlou, Vice President of Greek Lysosomal Association
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:
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