Contents

• 6th FIN Fabry Expert meeting, Lithuania
• Our Jack Johnson Receives PAL Award at WORLD 2018
• News from MetabERN
• Help raise funds for Fabry International Network with AVROBIO
• News From the Pharma Industry
• Remembering Christine Lavery
FIN Fabry Expert Meeting
Vilnius, Lithuania
8th & 9th June 2018

Places are still available for the Fabry Expert Meeting held in Vilnius, Lithuania.

Confirmed speakers:
Professor Atul Mehta
Dr Rick Steeds
Dr Saunder-Plassman
Dr Derralynn Hughes
Dr Jacqueline Adam
Martynas Davidonis
Prof Carolyn Elloway
Dr Aneal Khan
Prof Francois Eyskens
Plus many more……..

Further details and booking form available [here](#)
WORLDSymposium™ ANNOUNCES JACK JOHNSON AS RECIPIENT OF THE 2018 PATIENT ADVOCATE LEADER (PAL) AWARD

Each year, WORLDSymposium recognizes one individual for patient advocacy leadership in the field of lysosomal disease. The 2018 Patient Advocate Leader (PAL) award will be presented to Jack Johnson, founder of the Fabry Support & Information Group.

Jack is a founding member and Executive Director of the Fabry Support & Information Group (FSIG) in the United States, and is also a founding member and Vice President and Board Member of the Fabry International Network (FIN). It is the mission of the Fabry Support & Information Group to provide the Fabry community and the general public with information, advocacy, education, and compassionate support to improve the quality of life and the quality of care for Fabry patients and family members. Jack’s active role in the Fabry community on a national and international level has helped not only increase awareness for Fabry disease, but also has directly benefited numerous people who have been diagnosed with Fabry disease. Jack is able to bring his own experiences as a Fabry patient to his international advocacy work. Jack is a tireless advocate, and continues to champion patient meetings, fundraisers, and awareness activities for people affected by Fabry disease.

WORLDSymposium will honor Jack with the 2018 Patient Advocate Leader Award on Wednesday, February 7, 2018 at 7:45 AM prior to the start of the Translational Research presentations at WORLDSymposium 2018.

“Learning I was selected to receive the Patient Advocate Leader (PAL) Award at the 2018 WORLDSymposium was a real surprise. Barbara Wedehase, the former Executive Director of the National MPS Society in the USA and the late Christine Lavery, former Chief Executive of the MPS Society UK and President of FIN, were the first two award recipients. To me that was a no brainer. Being the third award recipient is an amazing honour and a vote of confidence that I have made a difference as well. I will keep doing my best to live up to what that means.”

Jack Johnson
News from Rare Disease Day 2018
Rare Disease Day takes place on the last day of February each year. It is an international event, which aims to create awareness about rare diseases and their effects on patients' lives. Let's get involved and create awareness for all rare disease patients.  
Read More

26th EURORDIS Round Table of Companies Workshop, 21 February 2018
The development of rare diseases therapies requires the right regulatory, economic and political ecosystem to ensure that investments are made in areas where research would not otherwise be carried out. This workshop will try to take a balanced look at the role of incentives in therapies development, address shortcomings of the current system and consider what else the rare disease community can do to fulfil important unmet medical needs.  
Read more

MetabERN

MetabERN held its first meeting with Patient and Family Associations
MetabERN held its first meeting with Patient and Family Associations on the 13th of January 2018. The meeting aimed to give an overview of MetabERN’s objectives for the future, to plan and define activities and programmes centred on high quality patient care, and to create a good basis for working together throughout the project.  
Download Report

New light shines on our understanding of Rare Diseases
French and Canadian scientists make discovery that could affect diagnosis, genetic counselling and therapeutic approaches in patients with a rare condition.
Rare hereditary recessive diseases were thought to be expressed in off-spring only when both parents carry a mutation in the causal gene, but a new study is changing this paradigm.  
Read More
AVROBIO is a clinical stage company developing disruptive ex vivo gene therapies that have the potential to transform patients' lives in a single dose.

AVROBIO is supporting Rare Disease Day with its ripples4rare campaign. Sharing your message of hope for rare disease will create a ripple. The more ripples we make, the more AVROBIO will donate to Fabry International Network. Share your message of hope today!

SHARING RIPPLES, MAKING WAVES

Every time you share this page you will help put us closer to our goal. Dedicated to all rare disease organisations, our focus this year is Fabry International Network (FIN), with a donation goal of $10,000.
News from

U.S. FDA Files New Drug Application Under Priority Review for Migalastat for Treatment of Fabry Disease

CRANBURY, N.J., Feb. 12, 2018 (GLOBE NEWSWIRE) -- Amicus Therapeutics (Nasdaq:FOLD) today announced that the U.S. Food and Drug Administration (FDA) has accepted the New Drug Application (NDA) for filing under priority review for the oral precision medicine migalastat HCl (“migalastat”) for the treatment of patients 16 years and older with Fabry disease who have amenable mutations. The Prescription Drug User Fee Act (PDUFA) goal date for the FDA decision is August 13, 2018.

Migalastat previously received both Orphan Drug Designation and Fast Track designation from the U.S. FDA. The FDA’s Priority Review status accelerates the review time from 10 months to a goal of six months from the day of acceptance of filing and is given to drugs that may offer major advances in treatment or may provide a treatment where no adequate therapy exists.

John F. Crowley, Chairman and Chief Executive Officer of Amicus Therapeutics, Inc., stated, “The FDA’s acceptance of our first Amicus NDA submission under priority review is an important step toward a potential oral precision medicine option for the Fabry disease community in the U.S. With more than a decade of experience in treating patients with migalastat globally, our team at Amicus has collaborated with leading Fabry disease experts and patient organization leaders to assemble a robust NDA that emphasizes the breadth of our clinical data and experience delivering migalastat to patients. We look forward to continuing to work collaboratively with FDA to bring this oral precision medicine to patients who may be able to benefit.”

The NDA submission for migalastat is based on clinical data from completed studies, including reduction in disease-causing substrate (GL-3), as well as the totality of data from two Phase 3 pivotal studies in treatment-naïve (Study 011, or FACETS) and enzyme replacement therapy (ERT) switch patients (Study 012, or ATTRACT).

An estimated 3,000 people in the U.S. are currently diagnosed with Fabry disease, more than any other country. Fabry disease is a progressive, inherited lysosomal storage disorder caused by deficiency of an enzyme called alpha-galactosidase A (alpha-Gal A), which is the result of mutations in the GLA gene. The disease causes accumulation of specific lipids, primarily GL-3, in tissues including the heart, kidneys, central nervous system, and skin. This abnormal accumulation can lead to debilitating consequences including pain, kidney failure, heart disease, and stroke.

The European Commission (EC) 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-Gal A deficiency) and who have an amenable mutation. The EC approval was based on clinical data from two Phase 3 pivotal studies (FACETS and ATTRACT), as well as ongoing long-term extension studies.

Outside the EU, migalastat is approved in Switzerland, Israel, Australia, South Korea, and Canada, with regulatory submissions under review in the U.S., Japan, and Taiwan.

About Migalastat and Amenable Mutations
Migalastat is a first-in-class chaperone therapy approved in the European Union as a monotherapy for Fabry disease in patients with amenable mutations. Migalastat works by stabilizing the body's own dysfunctional enzyme, so it can clear the accumulation of disease substrate in patients who have amenable mutations. A proprietary in vitro assay (Galafold Amenability Assay) has been used to classify more than 1,000 known GLA mutations as “amenable” or “not amenable” to treatment with migalastat. The EU label includes 348 GLA mutations that have been identified and determined to be amenable based on the Galafold Amenability Assay, which represent between 35% and 50% of the currently diagnosed Fabry population.
Healthcare providers in the EU may access the website www.Galafoldamenabilitytable.com to quickly and accurately identify which mutations are categorized as “amenable” or “not amenable” to migalastat. Amicus expects to submit additional updates to the EU label as additional GLA mutations are identified and tested in the Galafold Amenability Assay.

EU Important Safety Information
Treatment with Galafold should be initiated and supervised by specialists experienced in the diagnosis and treatment of Fabry disease. Galafold is not recommended for use in patients with a non-amenable mutation.

Galafold is not intended for concomitant use with enzyme replacement therapy.
Galafold is not recommended for use in patients with Fabry disease who have severe renal impairment (<30 mL/min/1.73 m²). The safety and efficacy of Galafold in children 0–15 years of age have not yet been established.

No dosage adjustments are required in patients with hepatic impairment or in the elderly population. There is very limited experience with the use of this medicine in pregnant women. If you are pregnant, think you may be pregnant, or are planning to have a baby, do not take this medicine until you have checked with your doctor, pharmacist, or nurse.

While taking Galafold, effective birth control should be used. It is not known whether Galafold is excreted in human milk.

Contraindications to Galafold include hypersensitivity to the active substance or to any of the excipients listed in the PRESCRIBING INFORMATION.
It is advised to periodically monitor renal function, echocardiographic parameters and biochemical markers (every 6 months) in patients initiated on Galafold or switched to Galafold.

OVERDOSE: General medical care is recommended in the case of Galafold overdose.
The most common adverse reaction reported was headache, which was experienced by approximately 10% of patients who received Galafold. For a complete list of adverse reactions, please review the SUMMARY OF PRODUCT CHARACTERISTICS.
Call your doctor for medical advice about side effects.
For further important safety information for Galafold, including posology and method of administration, special warnings, drug interactions and adverse drug reactions, please see the European SmPC for Galafold available from the EMA website at www.ema.europa.eu.

About Fabry Disease
Fabry disease is an inherited lysosomal storage disorder caused by deficiency of an enzyme called alpha-galactosidase A (alpha-Gal A), which is the result of mutations in the GLA gene. The primary biological function of alpha-Gal A is to degrade specific lipids in lysosomes, including globotriaosylceramide (referred to here as GL-3 and also known as Gb₃). Lipids that can be degraded by the action of alpha-Gal A are called "substrates" of the enzyme. Reduced or absent levels of alpha-Gal A activity lead to the accumulation of GL-3 in the affected tissues, including the central nervous system, heart, kidneys, and skin. Progressive accumulation of GL-3 is believed to lead to the morbidity and mortality of Fabry disease, including pain, kidney failure, heart disease, and stroke. The symptoms can be severe, differ from patient to patient, and begin at an early age. All Fabry disease is progressive and may lead to organ damage regardless of the time of symptom onset.

About Amicus Therapeutics
Amicus Therapeutics (Nasdaq:FOLD) is a global, patient-centric biotechnology company focused on discovering, developing and delivering novel high-quality medicines for people living with rare metabolic diseases. The lead program in the Amicus portfolio is migalastat, an oral precision medicine for people living with Fabry disease who have amenable genetic mutations. Migalastat is currently approved under the trade name Galafold™ in the European Union, with additional approvals granted and pending in several geographies. The lead biologics program in the Amicus pipeline is ATB200/AT2221, a novel, late-stage, potential best-in-class treatment paradigm for Pompe disease. The Company is committed to advancing and expanding a robust pipeline of cutting-edge, first- or best-in-class medicines for rare metabolic diseases.
Forward-Looking Statements
This press release contains "forward-looking statements" within the meaning of the Private Securities Litigation Reform Act of 1995 relating to the clinical development, regulatory approval pathway, and prospects and timing of regulatory submission and approval of our product candidates for the treatment of Fabry disease. Any express or implied statements contained in this press release that are not statements of historical fact, including interpretation of guidance given by the U.S. FDA, may be deemed forward-looking statements. The inclusion of forward-looking statements should not be regarded as a representation by us that any of our plans will be achieved in a timely manner or at all. Any or all of the forward-looking statements in this press release may turn out to be wrong and can be affected by inaccurate assumptions we might make or by known or unknown risks and uncertainties. For example, with respect to statements regarding the goals, progress, timing, and outcomes of discussions with regulatory authorities, actual results may differ materially from those set forth in this release due to the risks and uncertainties inherent in our business, including, without limitation, changes in FDA guidance for regulatory approval, risks regarding the FDA’s interpretation of our clinical trial results, including the risk that results from completed clinical trials that supported approval by regulators in other jurisdictions will not be sufficient for U.S. FDA purposes, the risk that the FDA will require additional studies or data, the potential that regulatory authorities, including the FDA, EMA, and PMDA, may not grant or may delay approval for our product candidate and the potential that we may not be successful in commercializing our product candidates for Fabry disease in Europe or any other country in which approval is ultimately obtained, if any. In addition, all forward-looking statements are subject to other risks detailed in our Annual Report on Form 10-K for the year ended December 31, 2016 and the Quarterly Report for the quarter ended September 30, 2017. The FDA guidance described in this release was given as of a specific date and the FDA could change its position on the clinical end points or other standards for review and/or approval. You are cautioned not to place undue reliance on these forward-looking statements, which speak only as of the date hereof. All forward-looking statements are qualified in their entirety by this cautionary statement, and we undertake no obligation to revise or update this news release to reflect events or circumstances after the date hereof.
“Personally, I know Christine for about 20 years, in which I have seen her grow. She took full advantage of all the opportunities she received from the MPS Society and she did not leave any challenge as a patent advocacy. She knew what she wanted to achieve and went fully for it. Many younger colleagues watched her because she was an inspiring example. Thank you for everything you have given everyone. Rest in peace.”
Lut De Baere, Director Fabry international Network

“FIN has lost its very competent, knowledgeable and devoted president. I will be forever grateful for her sharing of her passion and enthusiasm for Japan. Rest in peace, Christine.”
Anne Grimsbo, Chair of the Danish Fabry Patient Organization.

“Knowing and working with Christine for several years I learned to look up to her and her professional touch and resilience in everything she accomplished. Her legacy to the rare disease community and her example are something that will keep the rest of us going for years to come. On a personal level my favourite memories of Christine are the times she let loose and we shared wonderful moments and laughs together.”
Anna Meriluoto, Director Fabry international Network

“Early in my development as a Fabry disease non-profit organization leader I had the good fortune to meet Christine. I learned a great deal from her and always regarded Christine as a mentor. Her passing is a great loss to the global lysosomal storage disease community.”
Jack Johnson, CEO of FSIG

“As a new member of a global patient organization like FIN meeting and most importantly working with Christine was a great school for me. I certainly believe her knowledge will be greatly miss for all the lysosomal community.”
Mary Pavlou, Director Fabry international Network

Despite knowing Christine for a only a short time I still had the possibility to learn and enjoy the best of her qualities not only in a professional area but also as a wonderful person. It was nice to work with her due to her perfect understanding, great skills of motivation, knowing and being capable of doing her job well and teaching those who needed this. Her personality was filled with joy, fairness, good mood and love to people.”
Martynas Davidonis, Director Fabry international Network
IMPORTANT ANNOUNCEMENT FROM OUR CHAIRMAN

Dear Friends

Following the tragic and sudden loss of our founder and former CEO Christine Lavery MBE, we would like to give you an update on the current situation.

You will be aware that Bob Stevens has been appointed as the new CEO and it is no secret that this was Christine’s wish for the future of the Society, indeed she only reiterated her belief that this was the way forward to the Chairman just days before her untimely death.

You may also be aware that on the wishes of her family, a very small and intimate funeral was held on 15th January 2018. The intention was to then have a more public memorial event later in the year.

A memorial event is now being arranged as follows:

Date: 13th April 2018
Venue: Amersham, Buckinghamshire
Time: 2.00pm – 4.30pm

We can assure you that we have chosen a beautiful venue for such an event which is only a mile or so away from MPS House. A number of speakers will be invited to talk about the various aspects of Christine’s life. There will be light refreshments available including tea, coffee, cold drinks, cakes and scones etc.

Places are limited and we are anticipating a high demand of attendees. Should numbers exceed the venue’s capacity a draw will then take place. In order to be able to attend please apply by returning the slip below to mps@mpssociety.org.uk or post to the address above. Deadline for received applications will be 16th March 2018. Invitations will be dispatched in due course.

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Total Number of guests (max 4 per group)