EURORDIS Call for a European Year for Rare Diseases

Campaign for European Year for Rare Diseases 2019

“We, rare disease patient representatives from more than 600 patient groups, on behalf of the 30 million people affected by rare diseases in Europe, hereby call upon the EU and the Member States to support the designation of 2019 as the European Year for Rare Diseases. A European year will help raise awareness and encourage researchers to focus on these rare, mostly unknown, seriously debilitating and often life-threatening diseases, which affect children and adults, in their physical, mental, emotional and behavioural capacities.

We need solutions to address the health and social challenges that we face in our daily life; we need solutions to tackle the obstacles faced by researchers and by therapy developers; we need a European approach to overcome the chronically lacking critical mass of patients, data, experts, and resources; we need to involve all interested parties, including industry and decision-makers, in order to create the conditions for better health and social care for all, including the ones affected by diseases that leave little hope to patients and their families. We need a European Year for Rare Diseases.

The year 2019 will mark a turning point in the history of rare diseases as we will celebrate 20 years since the adoption of the EU Regulation on Orphan Medicinal Products, which has boosted orphan drug development; 10 years, since the Commission adopted its Communication on Rare Diseases: Europe’s Challenges and the Council its Recommendation on an action in the field of rare diseases, delineating a common strategy for rare disease patients. It will also be the time to take stock of the progress made by the Health for Growth and Horizon 2020 EU Programs and start operating in view of the new EU multi-annual financial framework 2020-2025”.

EURORDIS

Join the campaign to make 2019 the European Year for Rare Diseases.

www.eurordis.org/eyrd2019
International Fabry Disease Experts meet TAFD in Taiwan

Taiwan Association of Fabry Disease (TAFD) was privileged to receive the visit of two Fabry Experts: Dr Mehta from United Kingdom and Dr Schiffmann from United States.

Their attendance at the Chinese Medical Association Annual Conference on 27th June in Taipei represented a great opportunity to share with them the 2014 Fabry Disease Patient Medical Care and Living Qualities Study Report.

At the conference, President Catalina Yin introduced the Taiwanese Fabry Patient Association with its mission, Patient Care program, and an overview of past years health educational activities.

TAFD has completed the 2014 Fabry Disease Patient Medical Care and Living Qualities study and presented the findings to Dr Mehta, Dr Schiffmann and Dr Niu.

Dr Mehta and Dr Schiffmann were impressed with the results of the local patient survey and encouraged TAFD to publish their study in an International Journal.

The visit of two Fabry Experts in Taiwan has been motivating. Catalina Yin said "We are now even more hopeful as together we are looking at a brighter future".

TAFD, Taiwan Association of Fabry Disease
Fabry Support Group Australia 20th Anniversary Celebration Dinner

Fabry Support Group Australia (FSGA) held their 20th Anniversary Celebration Dinner on the 26th July 2014 at the Rendezvous Grand Hotel in Melbourne.

The night was a huge success with 65 people from all across Australia attending the dinner. Among the guests were old and new FSGA members, past and present FSGA presidents, nurses, doctors and pharmaceutical industry representatives – we even had a table of delightful teenagers. There were representatives from other Support Groups and people who have been supporters of our members who enjoyed getting to know what the FSGA was all about.

As a tribute to members no longer with us, we had a Remembrance Corner set up to light a candle or place a picture up while reflecting on our loved ones.

Lea was wonderful as our MC and kept the festivities going all night with lucky door prizes, raffles and a few speeches. We had one very special announcement to make during the evening - and that was to induct Megan Fookes; Director of FSGA, as an Honorary Life Member. This was kept under wraps and was a surprise to Megan, who through all her dedication, passion and hard work has helped the FSGA go from strength to strength.

In a speech delivered by FSGA Past President Lea Chant, Megan was acknowledged for her dedication and hard work. “Megan is a master networker, believe me I have watched her work a room - it’s an amazing site. She influences high profile people to agree with many of her terms when it comes to patient advocacy and promoting the wellbeing of the likes of you and me! Megan was not content to have us sit by and have treatment based on politicians’ terms, she made contact with the Fabry International Network to see what the treatment protocol was overseas and demanded changes here. Such was her strength of character and determination. FIN recognised the powerhouse and claimed her as their own. She now heads the National Alliance for Australians living with a rare disease; Rare Voices Australia Ltd employed as their Executive Director.

After the cutting of the cake by past and present presidents, the remainder of the evening was spent catching up with old and new friends. The FSGA Committee is to be commended for their contributions to a wonderful and memorable event.
The 11th Anniversary of the Slovenian Fabry Disease Patient Association

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The 10th Anniversary of the Center for Fabry Disease treatment in General hospital Slovenj Gradec, Slovenia

By Bojan Vujkovac, MD and Vesna Korat, RN

Slovenian Fabry disease patients association was founded in 2003 and has approximately 60 members. It is a member of FIN and Eurordis. The association’s task is to raise public awareness on rare diseases, patient status, drug supply and health care system organization. The association brings together patients, their family members and friends as well as the medical staff.

The Center for Fabry disease treatment in General hospital Slovenj Gradec was established in 2004. In 2003 an enzyme replacement therapy was implemented. Since 1991, 46 (1/50.000 citizens) Fabry disease patients were discovered, 8 of them died. By establishing a proper financial system and recognition of the special status the center has through years developed a model of a multidisciplinary approach where patients’ care is coordinated and holistic.

On Thursday, the 18th of September the celebration took place at the Youth cultural Centre in Slovenj Gradec. The celebration was primarily intended for Fabry disease patients, patients from other associations for rare diseases, hospital employees, citizens, politicians and the media. Mrs. Karmen Bizjak Merzel, a Fabry patient and a mother of a young patient with this rare disease gave a very personal and profound speech on behalf of all patients with Fabry disease. She also presented the book Fabry heart which was created in collaboration between the Fabry association and the Fabry center. The book was published in three languages (Slovenian, Croatian and English) and speaks about the personal experiences of patients and those who posed the foundation for our Center. The other keynote speakers at the ceremony were Mr. Andrej Možina, MD, President of the Medical Association, Prof. Borut Peterlin, MD, Head of the Working Group on Rare Diseases in the Department of Ministry of Health who outlined the problem of rare diseases in Slovenia and abroad, Mr.Andrej Čas, the mayor of Slovenj Gradec and Mr. Janez Lavre, MD, the Director of General hospital Slovenj Gradec gave a warm welcome to the participants. Head of Fabry center Mr. Bojan Vujkovac, MD talked about the beginnings of treatment and the management of Fabry disease through years. He encountered the importance of collaboration between the patient associations, the health care providers and the policy makers. At the end all the present gave a common notice that positive values, successful and fruitful cooperation between patients, doctors and nurses are needed for a good patient care. The celebration was also enlivened by the cultural program of children’s folklore group and a chamber trio.

The next day, on 19th of September an international, symposium on the topic of Fabry disease and other rare diseases was held in Ljubljana. About 100 participants attended, which is a lot, depending on such a specialised topic. Many renowned and recognized European experts in the field of treatment of Fabry disease and the leading representatives of the various disciplines and clinics at Clinical center Ljubljana and General hospital Slovenj Gradec were hosted as lecturers. The symposium was held in lively discussions and exchange of expert opinions. The meetings were organized in order to attract attention to a relatively small group of people, this is a rare disease patients. It is therefore to draw attention to the importance of rare diseases in the wider social context.
PRESS RELEASE - November 15, 2014

Amicus Therapeutics Announces Positive Phase 3 Data on Cardiac and Composite Endpoints from Fabry Monotherapy Study 012 at American Society of Nephrology

Statistically Significant Improvement in Key Cardiac Parameter in Patients Switched From Enzyme Replacement Therapies to Migalastat

CRANBURY, N.J. and PHILADELPHIA, Nov. 15, 2014 (GLOBE NEWSWIRE) -- Amicus Therapeutics (Nasdaq:FOLD), a biopharmaceutical company at the forefront of therapies for rare and orphan diseases, today announced additional positive data on important secondary endpoints from its second Phase 3 study (Study 012) of the oral small molecule chaperone migalastat HCl (“migalastat”) for Fabry disease. In a poster at the American Society of Nephrology (ASN) Kidney Week 2014, results were presented from Fabry patients with amenable mutations in Study 012 who switched from standard of care enzyme replacement therapy (ERT) to migalastat as their only therapy for Fabry disease. A slide presentation featuring these data is also available at http://ir.amicustherapeutics.com/events.cfm.

Data from the Fabry Registry indicate that the leading cause of death in patients is from cardiac complications. In Study 012, patients who switched from ERT to migalastat showed a statistically significant decrease from baseline to month 18 in left ventricular mass index (LVMI). LVMI is a measure of cardiac hypertrophy, an increase in the size of the heart that has been associated with an increased risk of cardiac events in Fabry patients.

"Cardiac disease represents a major cause of morbidity and mortality in Fabry patients that is not adequately addressed with currently available treatment," said Professor Ales Linhart, MD, PhD, a leading expert in the cardiac manifestations of Fabry disease from Charles University of Prague. "The data showing further regression of left ventricular mass in patients switched from ERT to migalastat in this study are very promising since in other cardiac conditions a reduction of cardiac mass typically translates to significant improvements in long term outcomes."

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<tr>
<th>Cardiac ECHO Parameters - Change from Baseline to Month 18*</th>
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<tr>
<td>Migalastat Baseline Mean (Mean, 95% CI)</td>
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<tr>
<td>Left Ventricular Mass Index (LVMI) (g/m²)</td>
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*Read in blinded manner in centralized lab every 6 months **Normal LVMI: 43-95 (female), 49-115 (male) ***Statistically significant (95% CI does not overlap zero)

Migalastat also demonstrated a favorable outcome in a composite endpoint of Fabry-associated clinical events, which are a defined set of morbidities or worsening in a Fabry patient’s renal, cardiac, or cerebrovascular status. Fabry-associated clinical events were observed in 29% (10/34) of patients who switched from ERT to migalastat and in 44% (8/18) of patients who remained on ERT.
Dr. Kathy Nicholls, nephrologist from Royal Melbourne Hospital and University of Melbourne, and investigator with 7 years experience in treating Fabry patients in clinical studies of migalastat, said "Fabry patients randomized to switch from ERT to Migalastat showed stability in renal function at least comparable to those who continued on ERT. The new data on cardiac mass and clinical events are encouraging. An oral therapy for Fabry disease is very attractive to patients."

Study 012 was designed to compare the safety and efficacy of migalastat to ERT in patients with Fabry disease who have amenable mutations. As previously reported, migalastat successfully met both co-primary endpoints of comparability to ERT on both key measures of kidney function in Study 012.

"These data are profoundly important," said John F. Crowley, Chairman and Chief Executive Officer of Amicus Therapeutics, Inc. "Migalastat is not oral ERT. It is a unique small molecule with a novel mechanism of action designed to restore enzyme activity in key cell types and organs of disease in Fabry patients with amenable mutations. Migalastat has previously shown a consistent and durable effect on substrate reduction and stabilization of kidney function. Now, for the first time, we are presenting data at ASN that demonstrate migalastat has a significant effect on a key cardiac manifestation of the disease. We intend to seek approval of migalastat in both the European Union and in the U.S. We are on track to hold our pre-submission meeting with European regulators this quarter, and expect to interact with the FDA in early 2015. Our goal is to make migalastat monotherapy available for all Fabry patients with amenable mutations as quickly as possible."

About Study 012

Study 012 was a Phase 3, open-label study that compared oral migalastat to standard-of-care enzyme replacement therapies (ERTs) for Fabry disease (Fabrazyme® and Replagal®). The study enrolled 60 patients (26 males and 34 females) with Fabry disease with amenable mutations in a clinical trial assay who had been treated with ERT for a minimum of 12 months prior to study entry. These patients were randomized 1.5:1 to switch to migalastat (36 patients) or remain on ERT (24 patients) for the primary 18-month treatment period, after which they were eligible to receive migalastat in a 12-month extension phase.

The co-primary outcome measures were the mean annualized changes in estimated glomerular filtration rate (eGFR) and measured (iohexol) GFR (mGFR) assessed by descriptive comparisons of migalastat and ERT over 18 months. Secondary outcome measures included cardiac function assessed by echocardiography, as well as a composite of Fabry-associated clinical events (i.e. renal, cardiac, or cerebrovascular).

Overall based on results from mutations tested in the GLP HEK assay, Amicus continues to believe that approximately 30% to 50% of the Fabry population have mutations that are amenable to migalastat.

About Amicus Therapeutics

Amicus Therapeutics (Nasdaq:FOLD) is a biopharmaceutical company at the forefront of therapies for rare and orphan diseases. The Company is developing novel, first-in-class treatments for a broad range of human genetic diseases, with a focus on delivering new benefits to individuals with lysosomal storage diseases. Amicus’ lead programs include the small molecule pharmacological chaperones migalastat as a monotherapy and in combination with enzyme replacement therapy (ERT) for Fabry disease; and AT2220 (duvoglustat) in combination with ERT for Pompe disease.

1Nicholls, et. al., American Society of Nephrology 2014. Migalastat and Enzyme Replacement Therapy Have Comparable Effects on Renal Function in Fabry Disease: Phase 3 Study Results.

2Mehta 2009
Forward-Looking Statements

This press release contains, and the accompanying slide presentation contains, “forward-looking statements” within the meaning of the Private Securities Litigation Reform Act of 1995 relating to preclinical and clinical development of Amicus’ candidate drug products, the timing and reporting of results from preclinical studies and clinical trials evaluating Amicus’ candidate drug products and the projected cash position for the Company. Words such as, but not limited to, "look forward to," "believe," "expect," "anticipate," "estimate," "intend," "potential," "plan," "targets," "likely," "may," "will," "would," "should" and "could," and similar expressions or words identify forward-looking statements. Such forward-looking statements are based upon current expectations that involve risks, changes in circumstances, assumptions and uncertainties. The inclusion of forward-looking statements should not be regarded as a representation by Amicus that any of its plans will be achieved. Any or all of the forward-looking statements in this press release may turn out to be wrong. They can be affected by inaccurate assumptions Amicus 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 and the potential goals, progress, timing and results of preclinical studies and clinical trials, actual results may differ materially from those set forth in this release due to the risks and uncertainties inherent in the business of Amicus, including, without limitation: the potential that results of clinical or pre-clinical studies indicate that the product candidates are unsafe or ineffective; the potential that it may be difficult to enroll patients in our clinical trials; the potential that regulatory authorities may not grant or may delay approval for our product candidates; the potential that preclinical and clinical studies could be delayed because we identify serious side effects or other safety issues; the potential that we will need additional funding to complete all of our studies and, our dependence on third parties in the conduct of our clinical studies. Further, the results of earlier preclinical studies and/or clinical trials may not be predictive of future results. With respect to statements regarding projections of the Company’s cash position, actual results may differ based on market factors and the Company’s ability to execute its operational and budget plans. 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, 2013. 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 Amicus undertakes no obligation to revise or update this news release to reflect events or circumstances after the date hereof. This caution is made under the safe harbor provisions of Section 21E of the Private Securities Litigation Reform Act of 1995.

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Source: Amicus Therapeutics, Inc.
Genzyme is delighted to announce that nine patient organizations were chosen to receive 2014 Patient Advocacy Leadership (PAL) Award grants for their innovative and exciting projects. The successful applicants came from Australia, Brazil, Croatia, Japan, Peru, the Netherlands, the U.S. and the U.K. A joint proposal from the U.S. and Canada was also selected for an award. Forty-five patient organizations from 23 different countries around the world submitted applications for this year’s PAL program, including first-ever proposals from Japan, Argentina, New Zealand, and Austria.

Ideas for new and innovative projects ranged from board training programs to disease awareness events that span national borders, to several smartphone apps designed to make it easier for patients and physicians to access information about rare diseases. Each proposal was unique and sought to meet the particular needs of their LSD community. The External Review Committee spent many hours reviewing the entire pool of applications and examining the merits of each project. Thank you to all of the patient organizations who participated in this program. For the many organizations that submitted PAL Award applications and did not receive funding, we encourage you to check the PAL Awards website periodically for updates and information on the next PAL Awards grant cycle.

We are deeply grateful to our External Review Committee members – Patricia Collins, Jean Campbell, Kimberly Goodrich, and Erik Tambuyzer – for the expertise, dedication and enthusiasm they brought to this project. The 2014 PAL Award recipients are listed below:

AUSTRALIA: Mucopolysaccharide and Related Diseases Society
BRAZIL: Regional Association of Patients and Families with Fabry disease
CROATIA: The Croatian Alliance for Rare Diseases
JAPAN: Fabry Disease Family and Patients Group
PERU: Association of Patients with Lysosomal Storage Disorders
NETHERLANDS: International Pompe Association
UNITED KINGDOM: The Cure & Action for Tay-Sachs (CATS) Foundation
U.S.: Fabry Support and Information Group
U.S. and Canada (joint proposal): National Niemann-Pick Disease Foundation and the Canadian Chapter of the NNPDF

www.genzymerarecommunity.com
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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Latest News
Information
Connection
Support

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The next FIN Newsletter will be out on 27th March 2015. Please send your articles and contributions by email to Nawel Van Lin no later than 20th March. We look forward to publishing and sharing your latest news with our International Fabry Community!

Season’s Greetings

FIN wishes you and all those close to you a joyous holiday season and a very happy New Year!