FSIG Connection
News from the Fabry Support & Information Group

FSIG marks 20th anniversary at Expert Fabry Conference

By Jack Johnson
Executive Director

At this year’s third annual FSIG Expert Fabry Conference held in San Diego, CA, we celebrated our 20th year of service to the Fabry community.

Thinking back, it’s hard to believe so many years have elapsed! The conference was another wonderful success, and it was a true pleasure and honor to share this milestone with all the attendees.

The conference started March 4 with a “Fabry 101” presentation, going over the basics. This was followed by a social mixer, which included dinner.

The morning of the 5th was filled with presentations from Fabry expert physicians, leading into an afternoon with updates from pharmaceutical company representatives. The end of the day was dedicated to a series of breakout sessions covering a broader range of topics.

The evening’s events concluded with a 20-year celebration dinner. To commemorate this achievement, Sanofi Genzyme played a video that highlighted my and FSIG’s role in the community.

Amicus Therapeutics and their Patient Advisory Board then presented me with a special achievement award. Caregiver awards were also presented, with the biggest applause going to winners of Best Doctor—Robert Hopkin of Cincinnati Children’s Hospital, and Best Medical Professional—Dawn Laney of Emory University.

MORE PHOTOS ON PAGE 2!

Hardworking and dedicated, FSIG executive assistant Connie Baldwin and executive director Jack Johnson are the icing on the cake for FSIG supporters and Fabry patients.

As always, the conference provided children’s activities.
Dear Friends,

Looking back on the last 20 years, we’ve made so many strides in our fight against “the dragon.” Most important has been the advent of enzyme replacement therapies, and of course, other new therapies are still being considered for FDA approval.

This year’s Fabry Expert Conference was such a wonderful experience for me—and a great opportunity to celebrate this milestone in our organization. The past 20 years have come with heartaches from losses, but I have also gained a great deal from the opportunities to meet so many of you wonderful, caring people. It has not been all easy, and I cannot adequately express my feelings for the honors, warm support and caring words of encouragement. I could not do it without all the support from all of you and my family. It’s all been for you, and from the bottom of my heart, I thank you.

Together, we can keep going.

Jack

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LETTERS TO THE EDITOR

Dear FSIG,

I just wanted to write you all to tell you I really enjoy reading the FSIG Connection. The update is really helpful and I love how there is a variety of information for several categories.

Thank you for everything you all do, it is greatly appreciated.

Thanks, Mercedes Schrank

Dear FSIG,

Just wanted to thank you for making it possible for us to attend the Expert Fabry Conference 2016!!!

We had no problem with our flights and the wheelchair assistance was great! The hotel was great— the view was beautiful—the food was delicious - I loved it when the chefs cooked just for me! Everyone was so helpful for us.

The speeches were really informative and I was amazed that a person with Fabry’s Disease could walk so far! It is always interesting to talk with other people with Fabry.

Thank you for sharing FSIG’s 20th anniversary with us.

Helen Kelleher
**Migalastat access expanded**

**PRESS RELEASE Excerpts**

Amicus Therapeutics has initiated a reimbursed expanded access program (EAP) to provide Fabry patients who have amenable mutations with access to migalastat. The EAP will be implemented in certain territories where reimbursement can be secured prior to marketing authorization and commercial availability, beginning in France.

The French National Agency for Medicines and Health Products Safety (ANSM) has granted an “Autorisation Temporaire d’Utilisation dite de cohorte” (cohort ATU), a Temporary Authorization for Use for patient sales of migalastat for Fabry patients who have amenable genetic mutations who meet the criteria for the cohort ATU.

An ATU is the regulatory mechanism used by the ANSM to make non-approved drugs available to patients in France when a genuine public health need exists. The ATU for migalastat was granted after an assessment by the ANSM of a full application dossier that included clinical and safety data from clinical studies of migalastat.

In France, patients with Fabry disease who have amenable mutations may receive treatment with migalastat before marketing authorization for the product is granted in the European Union. Government allocations to hospitals will allow payment for migalastat for patients included in the ATU program.

“We are grateful to the French authorities for granting an ATU to allow Fabry patients in France to begin treatment with migalastat and we are pursuing additional opportunities for early access in other countries.”

John F. Crowley, Amicus Chairman/CEO

**Migalastat oral treatment recommended for EU approval**

**PRESS RELEASE Excerpts**

The European Medicines Agency has recommended granting a marketing authorization in the European Union for Galafold (migalastat) for the treatment of Fabry disease.

Galafold is the first oral treatment for Fabry disease and may provide a more convenient treatment option for patients. Galafold is to be used only in patients with specific mutations of the disease which are known to be responsive to the active substance in the medicine, migalastat.

The evaluation of EMA’s Committee for Medicinal Products for Human Use (CHMP) was based on the results of two phase III clinical trials in about 110 patients with Fabry disease who had a genetic mutation which responds to migalastat.

The opinion adopted by the CHMP at its March meeting is an intermediary step on Galafold’s path to patient access. The CHMP opinion will now be sent to the European Commission for the adoption of a decision on an EU-wide marketing authorisation. Once a marketing authorisation has been granted, decisions about price and reimbursement will take place at the level of each Member State, taking into account the potential role/use of this medicine in the context of the national health system of that country.

**Positive data from PRX-102 early trials**

**PRESS RELEASE Excerpts**


PRX-102 is a recombinant plant cell expressed, Pegylated modified version of the human alpha-Galactosidase-A enzyme. Among trial findings were that participants exhibited stable cardiac and kidney function, as well as a meaningful reduction in the total score of Mainz Severity Score Index (MSSI), which looks at general, neurological, cardiovascular and renal parameters. View a PDF of the presentation at www.bit.ly/1NTNWHz.

**Phase III clinical trial for PRX-102 under way**

**PRESS RELEASE Excerpts**

Protalix BioTherapeutics, Inc., announced in November it recently held an End-of-Phase II meeting with the U.S. Food and Drug Administration (FDA) to discuss the company’s proposed BLA plan for PRX-102 for the treatment of Fabry disease. Official FDA meeting minutes indicate the FDA’s acceptance of the company’s path forward for a phase III clinical trial to support a full BLA approval.

The phase III clinical trial will be a randomized, multi-center, placebo-controlled, safety and efficacy study in treatment-naïve Fabry patients evaluating the 1 mg/kg dose of PRX-102. The primary endpoint will be gastrointestinal symptoms, with key secondary endpoints including renal function.

The FDA noted that the company reported interim analysis results from its phase I/II clinical trial of PRX-102 that preliminarily show a favorable trend in the severity and frequency of abdominal pain and frequency of diarrhea after six months of treatment with PRX-102—and that during a recent ERT shortage, patients who reduced or discontinued ERT dosing developed worsening of GI symptoms within a few weeks to months.
Get listed in the Fabry Registry!

What is the Fabry Registry?
The Fabry Disease Registry is a global resource dedicated to improving the understanding of the variability and progression of Fabry disease.

Any person with a confirmed diagnosis of Fabry disease is eligible to participate, regardless of disease type, treatment status or treatment choice. The Registry’s mission is to increase the understanding of Fabry disease to improve outcomes for patients with this disorder.

How Can I Participate?
Data Registry Services is making Registry participation as easy as possible for all interested individuals. Reach out to founders Eric Rice & Michelle Hackenberry directly, and they will facilitate the process. It’s simple and free. If you’re not sure if you’re already registered, they can answer that too. Contact: DATA REGISTRY SERVICES, LLC 412-877-1113 Eric@DataRegistryServices.com Michelle@DataRegistryServices.com

How Does It Benefit Me?
The data collected in the Registry is part of an ongoing observational study and it allows researchers and physicians to continually update their treatment options and the management of your disease.

Additionally, if you are participating in the Registry through Data Registry Services, we provide you with easy and free access to all your current and historical health records in one location.

How “Beyond the Diagnosis” exhibit debuts

Press Release Excerpts
The Rare Disease United Foundation created the Beyond the Diagnosis Art Exhibit to encourage the medical community to look beyond the diagnosis to the rare disease patient. Professional artists painted portraits of rare disease children which became part of a traveling exhibit for medical schools.

With an average diagnosis time of 8 years in a field where the parent is most often the expert, doctors and rare disease patients need to have a more engaged approach to improve quality of life. The Beyond the Diagnosis Art Exhibit was unveiled at Brown University’s Alpert Medical School for the entire month of February 2015. The Beyond the Diagnosis Art Exhibit will travel to Harvard Medical School in November 2015.

“There can be a lot of insecurity on the part of the physician or a medical student because you are supposed to be the smartest person in the room,” said Dr. Craig Eberson, a surgeon and Associate Professor of Orthopaedics at Brown University’s Alpert Medical School. He added that when he treats patients with rare diseases, it has to be in collaboration with the patient and the family. They are the experts. The Beyond the Diagnosis Art Exhibit leaves a lasting visual imprint and creates a unique connection to the viewer in a medical landscape where patients are too often misdiagnosed and misunderstood.

Better Data = Better Research = Better Treatment!

Putting Patients First

Basketball court named for popular coach with Fabry

Teacher received new heart 6 years ago, daughter says

By Kevin Haskin
The Topeka Capital-Journal

Before the tribute ended Tuesday night, it appeared Ben Meseke had called on Hayden to engage in a full-court press.

Several former basketball players and cross country runners joined the former Wildcat coach and his family on the floor inside the Bueltel Activities Center. The official naming of Ben Meseke Court happened at halftime of Hayden’s boys game against Topeka West.

“I taught a lot of classes, gave a lot of talks,” Meseke said of his time at the high school, “But this is going to be the toughest because this means so much to me. It is totally overwhelming.”

Meseke, who taught math for 23 years at Hayden and also served as assistant principal, directed the Wildcats from 1980-97, leading them to six Class 4A boys basketball titles in 12 state appearances. He also guided Hayden to five Centennial League titles while going 253-158. Meseke also coached Hayden to six state cross country championships.

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Patient Reported Databases Aid Drug Development

Patient-reported information in disease databases can help drugmakers to develop new therapies, especially for rare diseases, STAT News reports. Patients’ own reports provide a better sense of how their diseases and treatments affect them on a day-to-day basis, experts say, and also can provide companies with a better understanding of the potential demand for a new therapy before it hits the market.

“We’re all in this together,” Meseke said, “It’s been such a privilege to be there.”

Reprinted with permission. Full story: bit.ly/1Tlb3WZ
reason why he is there. But it’s hard for me not to view him as a specialist. Because that’s all I’ve known since I saw a pediatrician when I was younger!

I need someone to tell me whether something I’m experiencing is beyond “everyday symptoms of Fabry.” For example, I went in for a physical and a debriefing (since this is the second time I’ve seen him since I moved, and I’ve been having terrible vertigo and nausea).

It was easy for me to brush this off as “just another Fabry symptom”—except, it wasn’t. My sinuses were inflamed and my head was not draining, which left me dizzy, which caused the nausea. I didn’t even know I was sick! But I had been ignoring all the other signs because I’d passed them off as “frequent symptoms” instead of small alarm bells.

Lesson learned: It’s important to listen to your body and go over everything—no matter how small you feel it is—with a parent and/or doctor. They will tell you if it is something to look further into. Ignoring it because you’re “already diagnosed” can make you miss something... something that, when addressed, could improve all the other parts of you!

My advice is to be the specialist of you—the whole you. It’s the key to true wellness.

Amanda

DEAR DRAGON SLAYERZ,

It seems I have a specialist for everything! So much so, that when I finally went to the doctor (I moved recently), he asked me what he could possibly do for me that the other specialists could not. To be honest, I was stumped. After all, I have my own:

- Fabry specialists
- Geneticist who helps organize the tests
- Neurologist who tests the degeneration of my nerves
- Cardiologist to monitor my heart
- Gastroenterologist for my stomach and managing my IBS
- Nephrologist for my kidneys
- Ophthalmologist to keep an eye on my eyes
- Psychiatrist to monitor depression
- Pain doctor to manage the medication for my nerves

After explaining all of this to my new doctor, I sighed and just said, “I need to know what it all means together—I need someone for the whole of me.”

Physical wellness is not just the individual organs that make you up. I am the type to see a minute clinic if I feel I have a cold, instead of inconveniencing my primary doctor... even though that’s a good

Senate confirms Robert Califf as new FDA commissioner

PRESS RELEASE Excerpts

In January, U.S. Food and Drug Administration Commissioner Margaret A. Hamburg, M.D., appointed Robert Califf, M.D., a recognized global leader in cardiology, clinical research, and medical economics, as FDA Deputy Commissioner for Medical Products and Tobacco.

In this position, Dr. Califf will provide executive leadership to the Center for Drug Evaluation and Research, the Center for Biologics Evaluation and Research, the Center for Devices and Radiological Health and the Center for Tobacco Products. He will also oversee the Office of Special Medical Programs in the Office of the Commissioner. Dr. Califf will play a critical role in providing high-level advice and policy direction on the agency’s medical product and tobacco priorities and will manage cross-cutting clinical, scientific and regulatory initiatives in several key areas for the agency, including personalized medicine, orphan drugs, pediatric science and the advisory committee system.

“I am delighted to announce this important addition to FDA’s senior leadership team,” said FDA Commissioner Margaret A. Hamburg, M.D. “Dr. Califf’s deep knowledge and experience in the areas of medicine and clinical research will enable the agency to capitalize on, and improve upon, the significant advances we’ve made in medical product development and regulation over the last few years.”

Before he took office in late February, Dr. Califf was serving as vice chancellor of clinical and translational research at Duke University. Dr. Califf is recognized by the Institute for Scientific Information as one of the top 10 most cited medical authors, with more than 1,200 peer-reviewed publications.
Novel cell/gene therapy targets Fabry

PRESS RELEASE Excerpts
AVROBIO, a clinical-stage biotechnology 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. Jeffrey Medin (now at the Medical College of Wisconsin) at the University Health Network (UHN) in Toronto, ON.

Phase 1 programs will be in the clinic by early to mid-2016 in both acute myeloid leukemia (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.

Blood-brain barrier loosened to deliver medicine

By Laura Sanders
Science News

In its job protecting the brain from would-be invaders, the blood-brain barrier also blocks medicines from reaching the brain. But on November 5, ultrasound zaps shook loose that tight barrier in a woman who has a brain tumor, potentially granting entry to a chemotherapy drug. The technique, which relies on tiny bubbles set jiggling by ultrasound beams, has shown promise in animal models (SN: 9/27/08, p. 20). This is the first time it has been tried on a person, says neurosurgeon Todd Mainprize of Sunnybrook Health Sciences Centre in Toronto, who led the procedure.

Mainprize and colleagues injected microbubbles, a chemotherapy drug and an imaging agent that could be visualized by a scanner into the blood of the woman. Then, targeted ultrasound beams passed through her brain, where they made the microbubbles in her blood vessels contract and expand. This jostling temporarily opened the blood-brain barrier, allowing the imaging agent — and presumably the drug — to enter the brain tissue near her tumor, Mainprize reported in a November 10 media briefing.

The unpublished results are preliminary; the researchers don’t know how much of the drug made it into the tumor, or how the patient will fare long-term.

Mainprize and colleagues plan to perform the procedure on other patients to test whether the procedure is safe and feasible. If so, the method might ultimately be used to deliver medicine to treat a wide range of brain maladies such as tumors and Alzheimer’s disease. Reprinted with permission scien...
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