Senate passes bill to create national RD day

Press Release

WASHINGTON, D.C. – In February, the U.S. Senate passed a bipartisan resolution led by U.S. Sens. Sherrod Brown (D-OH) and John Barrasso (R-WY) that would recognize the last day of February as “National Rare Disease Day” to raise awareness of, and provide support for, the 30 million Americans living... See SENATE, page 3

FSIG helps Amicus ring NASDAQ closing bell

Press Release

Amicus Therapeutics, Inc. [FOLD], a biopharmaceutical company at the forefront of therapies for rare and orphan diseases, visited the NASDAQ MarketSite in Times Square in celebration of Rare Disease Day 2014. Rare Disease Day is held the last day of February each year and is the occasion to... See NASDAQ, page 3

Rare Disease Day less rare

Last day of February becoming internationally recognized

By Jack Johnson
FSIG Executive Director

The first Rare Disease Day, sponsored by EURORDIS, was held in Europe on February 29, 2008. February 29 was chosen because it is a rare day and it is symbolic of rare diseases. The first Rare Disease Day in the U.S. was observed in 2009, sponsored by the National Organization for Rare Disorders (NORD). The Rare Disease Day movement has spread around the world, and today is observed in many countries. It is always held on the last day of February. The importance of Rare Disease Day... See RARE DISEASE DAY, page 2

Genzyme marks day with events around the world

Press Release

CAMBRIDGE, Mass - Genzyme announced Feb. 28 its support of International Rare Disease Day with a series of initiatives meant to call attention to rare diseases as an important public health issue and to improve rare disease education, research, and treatment. February 28, 2014, marks the seventh International Rare Disease Day. This year’s theme, “Care: Join Together for Better Care,” emphasizes that the many different facets of rare disease care represent a universal need for patients and their families around the... See GENZYME, page 2

Genzyme expands facility

Page 6

Genzyme expands facility

Page 6

FDA off hook for shortage

Page 6

Brain scans offer pain hope

Page 7

Therapy works best for pain

Page 7

FDA limiting hydrocodone

Page 7

News briefs

Page 8

Dragon Slayerz Club

Page 9

Help stop bullying

Page 9

Boy’s “Wish” comes true

Page 9

Family’s Fabry fundraiser

Page 10

Orphan drug pioneers retire

Page 10

Genzyme opens PAL grant

Page 10

Acknowledgements

Page 11

Rare Disease Day less rare

Page 1

Genzyme marks Feb. 28

Page 1

Senate bill for national day

Page 1

FSIG, Amicus at NASDAQ

Page 1

FSIG Expert Conference

Page 4

New fund covers testing

Page 6

Genzyme expanding facility

Page 6

FDA off hook for shortage

Page 6

Brain scans offer pain hope

Page 7

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Page 7

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Page 7

News briefs

Page 8

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Page 9

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Page 9

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Page 9

Family’s Fabry fundraiser

Page 10

Orphan drug pioneers retire

Page 10

Genzyme opens PAL grant

Page 10

Acknowledgements

Page 11

1
Dear Friends,

We had a fantastic FSIG Expert Fabry Conference in San Diego this year with wonderful weather and are looking to do it again next year in Orlando!

Details are still in the works, but save the dates for February 13–15, 2015. We will be providing more information as soon as it is set.

The Fabry world has a lot happening and unfortunately, due to space constraints, we could not report on all of it. But a good cross section of information is included in this release of FSIG Connection. The future holds excited potential for improved understanding of Fabry disease as well as improvements in treatment and care.

Thanks for reading,

Jack
Jack Johnson
FSIG Executive Director

FSIG would like to express our gratitude to the many physicians, health care professionals, researchers, scientists, and industry working on our behalf. Their efforts are not always evident but make a great difference for us all.

GENZYMЕ, continued from page 1

world. Advocating at the local and national level to improve the lives of people living with a rare disease and their families is necessary.

Observed annually on the last day of February, Rare Disease Day is an international awareness campaign organized by the patient organization EURORDIS and supported by hundreds of other patient organizations worldwide.

Genzyme sites around the world are partnering with local patient organizations on a variety of Rare Disease Day activities meant to educate policymakers, medical professionals, patients and caregivers, and the general public and empower them to advocate for those affected by rare diseases. These initiatives include:

- **Netherlands:** In Naarden, over 50 employees join together for the “Express That You Care” workshop, creating 3 colorful paintings—one of the paintings will move to the new building of the Dutch Association for Children and Parents with Metabolic Diseases (VKS).
- **Japan:** Genzyme is the primary sponsor of Japan’s main Rare Disease Day event, held at Tokyo Station. The event includes a panel exhibition, speeches about rare diseases from patients and students, and a library of rare-disease-themed books.
- **Italy:** Genzyme is partnering with the Italian Rare Disease Patient Movement and the Italian Glicogenosis Association to showcase photos of people living with rare diseases in the Fotograf Rare exhibition.
- **Spain:** Genzyme is working with the Federation for Rare Diseases (FEDER) to teach schoolchildren about the daily realities faced by their classmates living with rare diseases through an ongoing program called “Take a Rare Challenge.”
- **Brazil:** In São Paulo, Genzyme employees join patients and other supporters for a Rare Diseases Street Walk between Mario Covas Park and Trianon Park.
- **Philippines:** Genzyme is working with the Philippine Society for Orphan Disorders, Inc. (PSOD), to hold a Household Business Forum teaching caregivers of rare disease patients how they can earn a living while staying home to care for their family member.
- **Germany:** Healthcare policies related to this year’s theme “caring” will be discussed with an interdisciplinary “expert talk round” and with journalists.
- **Hong Kong:** Genzyme is partnering with the Hong Kong Alliance of Patients’ Organizations (HKAPO), the Hong Kong Mucopolysaccharidoses and Rare Genetic Diseases Mutual Aid Group (HKMPS), and various stakeholders to organize a roundtable on rare disease policy development. HKAPO will use the output of the meeting to develop a white paper report on rare disease policy in Hong Kong.
- **France:** Genzyme has joined forces with the patient association Alliance Maladies Rares to explain rare diseases to children by creating a partnership with the newspaper “Mon Petit Quotidien.” The proposed program is developed for French schools.
- **From Australia to the UK:** Genzyme is sponsoring a Rare Disease Day running/walking relay which connects 12 Genzyme sites around the world.

RARE DISEASE DAY, continued from page 1

has also grown in importance across the U.S., with more organizations becoming involved every year.

This year, FSIG took a more active role to help spread public awareness when I joined the efforts of Tina Croghan and Mary Schultz from the Spastic Paraplegia Foundation.

On Friday, February 28, we met at the Missouri State Capitol in Jefferson City. This was the state’s first Rare Disease Day event, and we shared information about the importance of rare diseases in general and about the conditions each of our organizations represent.

A table was set up in the Capitol Rotunda, where information was provided to those visiting the Capitol and museum exhibits. It turned out to be a fairly quiet day for visitor traffic, so information was spread to state representatives’ staff and others in a door-to-door stroll through the building. In this way, we spread awareness to many who were unaware of Rare Disease Day and had no knowledge of Fabry disease or Spastic Paraplegia, a condition related to Lou Gehrig’s disease.
raise awareness for rare diseases. Amicus was joined by representatives from several patient advocacy organizations including the Global Genes Project, the National Organization for Rare Disorders, the Acid Maltase Deficiency Association, the Fabry Support and Information Group, the National MPS

SENATE, continued from page 1

with a rare and potentially life-threatening disease or disorder.

“Every day 30 million Americans, including too many children, have to live with life-limiting and sometimes life-threatening rare diseases,” Brown said. “National Rare Disease Day is an opportunity for Congress to bring attention to these brave Americans, increase awareness about rare diseases, and help us understand the need for advances that can lead to prevention, effective treatments, and cures.”

There are a combined 7,000 rare diseases that affect nearly 30 million Americans. More than half of these Americans are children, and many of these rare diseases are serious and life-threatening. Because these diseases are rare, patients often encounter delays and obstacles obtaining an accurate diagnosis, limited treatment options, and difficulty finding physicians or treatment centers with expertise in their diseases. The resolution recognizes the importance of both the Food and Drug Administration (FDA) and the National Institutes of Health (NIH) in establishing special offices to advocate for rare disease research and treatments, and the National Organization for Rare Disorders for advocating on behalf of patients with rare diseases.

Examples of life-threatening rare diseases include: epidermolysis bullosa; progeria; sickle cell anemia; spinal muscular atrophy (SMA); Duchenne muscular dystrophy (DMD); Tay-Sachs; cystic fibrosis; pulmonary fibrosis, many childhood cancers; and fibrodysplasia ossificans progressiva.

Brown continues to fight on behalf of Americans fighting rare diseases. Last year the U.S. Congress passed, and President Obama signed into law, the National Pediatric Research Network Act, bipartisan legislation Brown authored that created a more streamlined and efficient system for maximizing pediatric medical research—with an emphasis on rare pediatric diseases.

The law is endorsed by the American Academy of Pediatrics, American Board of Pediatrics, American Pediatric Society, Association of Medical School Pediatric Department Chairs, Association of Pediatric Program Directors, Children’s Hospital Association, Coalition for Pediatric Medical Research, Federation of Pediatric Organizations, FightSMA, EveryLife Foundation for Rare Diseases, National Down Syndrome Society, Parent Project Muscular Dystrophy, and Society for Pediatric Research. 

NASDAQ, continued from page 1

RARE DISEASE DAY

In honor of the occasion, John Crowley, Chairman and CEO of Amicus rang the Closing Bell.
and learning, everyone came back together for an evening of awards, fun and networking. Awards were given to four individuals nominated for the Best Caregiver of the Year. These went to Jacqui Howells (first) and three runners-up: Sabina Kaneen, Karen Rohrs and Pam Fuller. Geoffrey A. Block, M.D., won the Best Doctor Award, and Renee Dickerson, R.N., received the Best Medical Professional Award. A 50/50 raffle was also held, and everyone enjoyed great laughs courtesy of Milo Shapiro and an improv group.

The conference wrapped up Sunday morning with two informal focus group-style sessions combining “Caregivers with Females & Fabry” and “FSIG and You.” A trip to the San Diego Zoo gave a final opportunity to enjoy the wonderful weather and see some local sights before everyone headed home.

Based on comments and responses to the evaluation forms, the first FSIG Expert Fabry Conference was a true success. We will be utilizing comments to make future events even better.

By Jack Johnson
FSIG Executive Director

On February 14–16, FSIG followed the WORLD Symposium™ with the first FSIG Expert Fabry Conference—taking advantage of immediate access to the most up-to-date information and availability of top Fabry experts combined with San Diego’s wonderful weather proved a winning combination.

The first WORLD Symposium™ was held in Minneapolis on February 13, 2004, with about 100 physicians and researchers in attendance. Several lysosomal storage disease (LSD) advocacy groups, including FSIG, held meetings for area patients and family members the following day. I was fortunate to attend this inaugural event, and since this humble beginning, the WORLD Symposium™ has grown to become the premier LSD event, bringing together expert physicians, researchers and thought leaders from around the world.

The FSIG conference began on Friday, February 14, with registration, followed by a welcome dinner and social time. It was so wonderful to reunite with familiar faces and to meet new people. The majority of attendees were from the southern half of California and surrounding states, but others came from the Midwest and even East Coast states, including Jerry Walter with the National Fabry Disease Foundation (NFDF). The person who traveled the farthest, though, was a representative from the Japan Fabry Disease Patients and Family Association—our only international attendee, traveling nearly 6,000 miles!

During the morning and afternoon educational sessions, kids and teens had a great time in “The Hangout”—a room set aside just for the youth, where they enjoyed projects, games and a special guest named Abigail Carbone. Eighteen-year-old Abigail recognized a specific need for information targeted to tweens and teens, so she wrote a fantastic book titled The Long Road to Fabry.

She gave a personal reading for the group and then signed copies of her book for all the kids to take home. Abigail’s book was also provided to all conference attendees who wanted a copy. If you are a member of FSIG and interested in receiving a copy, just call or email us (info@fabry.org) and one will be sent free of charge.

After a long hard day of listening, sharing and learning, everyone came back together for an evening of awards, fun and networking. Awards were given to four individuals nominated for the Best Caregiver of the Year. These went to Jacqui Howells (first) and three runners-up: Sabina Kaneen, Karen Rohrs and Pam Fuller. Geoffrey A. Block, M.D., won the Best Doctor Award, and Renee Dickerson, R.N., received the Best Medical Professional Award. A 50/50 raffle was also held, and everyone enjoyed great laughs courtesy of Milo Shapiro and an improv group.

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The plan for FSIG’s first national conference was based on a survey we distributed at the end of August 2013. The meetings survey included questions about topics of importance to you—the Fabry community—and while not all topics could be covered, the highest-ranking responses were included.

We also asked about attending a national conference and limiting factors. Based on survey responses, we created the agenda and attendance estimate. Fewer than 100 participants were expected, due to finances and travel being listed as major limiting factors. Accordingly, we chose a hotel near the WORLD Symposium™ and set the budget.

To our surprise, the huge response to our conference announcement exceeded our wildest expectations. The hotel contract was expanded (twice!) for the growing attendance. We ultimately found ourselves limited by the number of people the Embassy Suites could accommodate in their largest meeting room.

While this remarkable level of interest was thrilling, the meeting room size limitation regretfully meant not everyone who wanted to attend was able to. But due to a number of cancellations, most did get the opportunity.

Behind the event: FSIG used your ideas!

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Sharing what we learned together

By Jack Johnson
FSIG Executive Director

With nearly 200 in attendance at the conference, experts in the treatment and care of Fabry disease gave detailed presentations Saturday morning. Michael Mauer, M.D., went over kidney involvement and biopsy studies that showed kidney damage starts very early. He explained what damage occurred in the kidneys, emphasizing that treatment dose matters and early treatment has potential long-term benefits.

Robert Hopkin, M.D., gave an overview of new research findings, which was the most requested topic from the meetings survey. Addressing numerous questions throughout his presentation, Dr. Hopkin started with a general overview of Fabry signs, symptoms and current treatment options. He touched on areas where additional research is still needed, and went through new forms of treatment that are in development either in clinical trials or still pre-clinical stages.

Dawn Laney, M.S., provided a presentation on the many research programs occurring at Emory University, and Raphael Schiffmann, M.D., covered pain issues related to Fabry, its mechanism and primary methods for treatment. Behzad Najafian, M.D., gave a second presentation on Fabry and the kidneys, but from a pathologist point of view—sharing information on noninvasive biomarker studies trying to better assess disease progression.

These talks were followed by presentations from Genzyme (a Sanofi Company), Amicus Therapeutics and Protalix Biotherapeutics, with updates on production, current and coming clinical trial programs, and data from clinical trials as well as laboratory work. All three companies have exciting things in the works. The afternoon was filled with breakout sessions, giving attendees an opportunity to spend more time on specific topics of interest and get questions addressed in a group setting for greater interaction with speakers and other attendees.

Chester Whitley, M.D., has been the main driving force for the WORLD Symposium™, and I recall him stepping in for the Fabry meeting we held 10 years ago. On this anniversary, he once again was able to participate in two breakout sessions on “Nutrition & Exercise,” along with Patrick Sorgen, PharmD, Jeanine Utz, PharmD, and Fabry patient Tim Falencik.

Connie Kreps, R.N., presented in breakout sessions on “Feeling Overwhelmed & Balancing Family” as well as “Living with Fabry Self & Family Members.” A panel-format discussion addressed questions in the “Ask Industry” breakout, with representatives from all the drug companies, and a panel of physicians and medical professions was available for an “Ask the Experts” breakout session on Fabry treatment.

A number of findings presented at the WORLD Symposium™ were covered, as well as some that were not at the symposium. These talks were followed by presentations from Genzyme (a Sanofi Company), Amicus Therapeutics and Protalix Biotherapeutics, with updates on production, current and coming clinical trial programs, and data from clinical trials as well as laboratory work. All three companies have exciting things in the works. The afternoon was filled with breakout sessions, giving attendees an opportunity to spend more time on specific topics of interest and get questions addressed in a group setting for greater interaction with speakers and other attendees.

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New NORD, Genzyme fund to cover diagnostic testing

Press Release
DANBURY, Conn.—Genzyme and the National Organization for Rare Disorders (NORD) announced in December the creation of a fund to pay for standard diagnostic testing for people with mysterious, undiagnosed medical conditions.

The fund will help those who have applied to the National Institutes of Health (NIH) Undiagnosed Diseases Program, but who cannot afford the basic medical tests needed to make them eligible to participate in the NIH program. “While progress in scientific research has been very promising in recent years, millions of Americans who have rare diseases go for five years or longer without an accurate diagnosis,” said NORD President and CEO Peter L. Saltonstall. “This delays treatment and creates enormous financial and emotional stress for patients and their families.”

Genzyme has been a longstanding partner of NORD, and the creation of the Genzyme/NORD NIH Undiagnosed Diseases Fund is another way for Genzyme to support the rare disease community. The new fund is made possible in part by a team of Genzyme employees who run the Boston Marathon® to raise funds for NORD services on behalf of all patients and families affected by rare diseases.

Expansion will up Fabrazyme® supply

FDA let off the hook for drug shortage

Excerpts from Courthouse News

A federal judge has ruled that The Food and Drug Administration won’t have to face claims that it helped create a shortage of Fabrazyme, a treatment for life-threatening Fabry disease.

A group of users claimed in a February 2012 complaint that the government had given the drugmaker a pass to create a Fabrazyme shortage. Genzyme was not named as a defendant to the complaint, which was lodged against the FDA, the Department of Health and Human Services, the National Institutes of Health, the Mount Sinai School of Medicine, and top officials in the federal agencies.

The 25 plaintiffs, 13 of whom sued anonymously, claimed that the shortage denied them interstate access to Fabrazyme.

But U.S. District Judge Beryl Howell dismissed the action just before Thanksgiving, saying that it was trying “to obfuscate the relevant facts of this matter, as it mixes facts, legal arguments, and political and social theory, often in the same paragraph.”

“As the defendants point out ‘[t]he precise nature of the claims in plaintiffs’ ... complaint is often difficult to discern,’” Howell said. Lead plaintiff Joseph Carik has a doctor’s prescription for Fabrazyme, and claimed that “FDA consent” banned him from interstate access to FDA-approved doses of Fabrazyme during the Genzyme-created drug shortage.

“However, the court concluded that the plaintiffs failed to demonstrate how the relevant facts of the claims in plaintiffs’ ... complaint is often difficult to discern,” Howell said. Lead plaintiff Joseph Carik has a doctor’s prescription for Fabrazyme, and claimed that “FDA consent” banned him from interstate access to FDA-approved doses of Fabrazyme during the Genzyme-created drug shortage.

“Issues raised by plaintiffs do not make the case more difficult to discern,” Howell said. “Notwithstanding the defendants’ assertions to the contrary, plaintiffs have failed to make the showing required by our Federal Rules of Civil Procedure. Although the precise nature of the claims in plaintiffs’ complaint is often difficult to discern, there is no showing that the FDA consented to prevent interstate access or that the consent was required by the shortage.”

“Under those circumstances, the plaintiffs are not entitled to an order directing the defendants to submit to an evidentiary hearing,” Howell said. “Instead, the defendants are entitled to dismissal based on the pleadings.”

Genzyme plans $80M processing facility

Press Release
CAMBRIDGE, Mass.—Genzyme, a Sanofi company, announced in October that it will be investing $80 million to build a new downstream processing facility for Fabrazyme® (agalsidase beta). The new plant, which will be located adjacent to the new Fabrazyme cell culture manufacturing site in Framingham, Mass., will significantly expand purification capacity to support anticipated growth in global demand over the coming years.

“As we continue to meet the global demands for Fabrazyme and build inventory, our focus also remains on the future needs of the global Fabry community,” said Genzyme President and CEO David Meeker, M.D. “Following last year’s regulatory approvals of our manufacturing facility in Framingham and a new vial filling line at our plant in Waterford, Ireland, we continue to execute on our global manufacturing strategy, enhancing our capabilities across the entire manufacturing process for Fabrazyme.”

Downstream processing, a vital step in biologic production, involves the purification of material harvested from the cell culture manufacturing process. The final product, which is administered as an intravenous (IV) infusion, is formulated and fill-finished in a separate facility in Waterford, Ireland and shipped to multiple distribution centers for labeling, packaging and shipping around the world.

Source: www.courthousenews.com/2013/12/03/63383.htm
Brain scans offer hope for new pain treatment

By University of Michigan Health System

ANN ARBOR, Mich. – A study in the December issue of Anesthesiology suggests a role for brain imaging in the assessment and potential treatment of chronic pain.

Brain imaging allows researchers to see pain medicines at work in fibromyalgia patients. University of Michigan researchers are the first to use brain imaging procedures to track the clinical action of pregabalin, a drug known by the brand name Lyrica that is prescribed to patients suffering from fibromyalgia and neuropathic pain.

Three different brain imaging procedures were performed, in 17 patients with fibromyalgia.

Patients with fibromyalgia may spontaneously report pain throughout their bodies although there is no inflammatory or anatomical damage. In addition to chronic pain, patients may also suffer from related mood disturbances, such as anxiety and depression.

Previous research has shown that fibromyalgia patients may have heightened neural activity in a region of the brain involved in processing pain and emotion called the insula, and that this excess activity may be related to elevated levels of the excitatory neurotransmitter glutamate.

Brain imaging conducted at the U-M Health System suggests pregabalin works in part by reducing the concentration of glutamate within the insula, which is consistent with animal studies. These reductions in glutamate were also accompanied by decreases in insula connectivity and reductions in clinical pain ratings.

This type of brain activity imaging may help in the development of new pain medicines and personalized chronic pain treatment.

“The significance of this study is that it demonstrates that pharmacologic therapies for chronic pain can be studied with brain imaging,” says lead study author Richard Harris, Ph.D., assistant professor of anesthesiology at the University of Michigan.

“The results could point to a future in which more targeted brain imaging approaches can be used during pharmacological treatment of chronic widespread pain, rather than the current trial-and-error approach.”

Source: http://tinyurl.com/mh7rsm7

Therapy often better than meds for chronic pain

From PsychCentral.com

According to an article on PsychCentral.com, more than a third of all Americans have some form of chronic pain, and that the pain is often inadequately controlled.

It reported that a new comprehensive review shows that psychological interventions often provide more relief to sufferers of chronic pain (and without the risk of side effects) than do prescription drugs or surgery.

Despite the documented benefit, the article continues, therapy is used much less frequently for pain relief than traditional medical treatments, as was shown in a comprehensive review published by the American Psychological Association.

“Chronic pain affects 116 million American adults, making it more prevalent than heart disease, diabetes and cancer combined, and traditional medical approaches are inadequate,” said Mark P. Jensen, PhD, University of Washington, the lead scholar for the review. Other articles in a special February issue of the APA's flagship journal, American Psychologist, describe how psychology addresses racial and ethnic disparities in the assessment and treatment of chronic pain, persistent pain in older adults and family influences on children’s chronic pain.

The articles also cover a range of successful treatment approaches for chronic pain, including cognitive-behavioral therapy, acceptance and commitment therapy, mindfulness, and hypnosis. Additional articles look at how neurophysiology can help tailor treatments for specific cases and how interdisciplinary chronic pain management is most likely to lead to effective outcomes when health care teams include psychologists and coordinate services.

FDA pushes new limits on painkillers with hydrocodone

Compiled from News Reports

In October, the Food and Drug Administration recommended tighter controls on prescription painkillers containing hydrocodone, in an effort to crack down on medication abuse. The news was met with mixed reactions.

ABC World News (10/24, story 3, 0:30, Sawyer) reported that the agency’s “plan would allow fewer refills. After three months, patients would be forced to go back to their doctor before getting more medicine.” Also, patients would have to take prescriptions to a pharmacy, and physicians would not be allowed to phone them in. The National Community Pharmacists Association reportedly said that the FDA decision will likely “pose significant hardships for many patients and delay relief for vulnerable patients with legitimate chronic pain.”
NEW NEWS BRIEFS

New MRI technique can work to diagnose Fabry

A new MRI technique developed at the University of Alberta has resulted in updated clinical guidelines for the diagnosis and treatment of Fabry in Canada. The technique, known as T1 mapping, can detect heart damage and changes at early stages—earlier than regular MRI scans or ultrasound. When this type of MRI is used on patients with Fabry disease, the scans can detect both the disease and the severity of damage to the heart.

“This test can uniquely identify Fabry disease by detecting microscopic changes in the heart muscle structure that are not visible on regular images,” said researcher Richard Thompson, who works in the Department of Biomedical Engineering.

Co-researcher Gavin Oudit added, “It is very likely that this technique will become a key part in clinical examination of patients with Fabry disease. This finding will advance the clinical care of these patients around the world. The implications will be widespread.”

Crisis: Chronic-condition patients skipping meds

An October report by a coalition of medical and consumer groups concludes poor adherence to medications among patients with multiple chronic conditions has reached “crisis proportions” in the U.S. It leads to “unnecessary disease progression” and complications and too many emergency room visits, hospitalizations, and avoidable hospital readmissions, says the National Council on Patient Information and Education.

Caring for the more than a quarter of Americans with multiple chronic conditions accounts for 66% of the nation’s health expenditures and is a major source of Medicare spending, the report concludes.

Among the solutions recommended by the coalition were lowering or eliminating co-payments for medications used to treat the most common chronic diseases. The group is also pushing pharmacies to better coordinate patients’ medications, among several other recommendations.

Risk of heart disease death correlated to Fabry

A study recently published in The Journal of the American Heart Association found that the risk of death in heart disease is associated with elevated urinary globotriaosylceramide, considered a hallmark of Fabry disease—which in itself is a risk factor for most types of heart disease.

The researchers screened 1421 consecutive patients with common forms of heart disease for Fabry disease by measuring urinary Gb3 in whole urine. Their conclusion was that, in heart disease patients who do not have Fabry disease or GLA gene mutations, a higher level of urinary Gb3 is positively associated with near-term mortality.

The elevation of urinary Gb3 and that of other lipids suggests that heart disease is associated with multigorgan lipid abnormalities.

Drinkable ERT successful for Gaucher sufferers

Protalix Biotherapeutics recently announced that PRX-112—the oral formulation of its injectable Elelyso for Gaucher disease—was well-tolerated, detectable in blood samples after administration, and biologically active in a phase 1 trial. If successful, such a treatment could challenge the current three injectable drugs from Protalix, Shire, and Sanofi (Genzyme), which dominate the market.

The unique ingredient of the therapy is genetically engineered carrot cells, which express the enzyme at the heart of the therapy and naturally encapsulate it within their plant cell walls.

Since plant cell walls are composed of cellulose, the enzyme is shielded from the digestive tract and can successfully pass and attach to the mucosal lining of the intestine. If successful, similar technology could be used to develop oral formulations of other ERTs for other diseases, including Fabry.

Amicus finishes Fabry study, plans next phases

Amicus has completed a Phase 2 clinical study of migalastat HCl co-administered with currently approved ERTs for Fabry disease (Fabrazyme® and Replagal®) as well as preclinical studies of migalastat HCl co-formulated with a proprietary investigational ERT for Fabry disease (JCR Pharmaceutical Co Ltd’s JR-051).

JR-051 is a human recombinant alpha-Gal A enzyme that is designed to be biosimilar to Fabrazyme. Positive results from these clinical and preclinical studies demonstrated increased enzyme activity in plasma and greater enzyme uptake into tissues in the presence of the chaperone compared to any of these ERTs alone.

FABRY WEBINAR

On January 21, Amicus Therapeutics and Patient Services Inc (PSI) sponsored an online educational webinar for the Fabry and Pompe communities to discuss PSI services and the Affordable Care Act. If you missed this webinar or just want to hear it again, listen at: http://tinyurl.com/l85f2b4

FOLLOW US! LIKE US!
DEAR DRAGON SLAYERZ,

I hope everyone is doing well. It was great to see so many kids show up back in February at the FSIG Expert Fabry Conference. To have that much support was a great thing to see among young Fabbers. I hope those who went got each other’s contact information for the future.

I cannot over-emphasize the importance of keeping a support system around. It will help with possible depression and keep you updated on new developing information on Fabry. If there is no one immediately around that you feel you can talk to, I urge you to email or call FSIG, and we will make sure you do not feel alone.

I hope you all stay cool this summer and remember to use spray bottles and wet towels if you are in direct heat!!

Looking forward to seeing you all again!!

Amanda

Help stop bullying!

Research shows that children and teens with disabilities and other special healthcare needs may be at increased risk of being bullied due to a number of factors, including being physically weakened, challenges with social skills, or intolerant environments.

Bullying can make a big impact on self-esteem and emotional health, for yourself or others you might see getting bullied.

Check out these sites focused on helping teens prevent bullying:

- PacerTeensAgainstBullying.org
- StopBullying.gov/kids
- TeenCentral.net/Bully

By Stacey Delikat
FOX 5 News, New York

NEW JERSEY - Children fighting terminal illnesses have seen some of their dreams come true through the Make-A-Wish Foundation. The organization recently helped one young Giants fan experience an unforgettable moment.

Jackson Kondak, a 9-year-old boy from Maplewood, New Jersey, had never been to a professional football game until he caught the New York Giants in action at MetLife Stadium through the Make-A-Wish Foundation. But that was just a part of his special experience with the team.

The boy suffers from Fabry, a rare genetic disorder that requires he spend five hours on any given day hooked up to an IV receiving enzyme infusions.

His mother Kim also suffers from Fabry. “It causes cell damage throughout the body,” she explained. “It’s a progressive disease, so kidney failure, heart failure, and strokes are part of Fabry.”

Every other Friday, Jackson and Kim spend five hours hooked up to IVs getting special enzyme infusions to slow the onset of Fabry.

But the treatment on Friday, Nov. 15, was rescheduled so he could have a once-in-a-lifetime experience, coordinated by Make-A-Wish of New Jersey.

“Our mission is very simple—it’s one sentence: We grant wishes to children with life threatening medical conditions to enrich the human experience with hope, strength and joy,” said CEO Tom Weatherall.

Jackson and his younger brother Christopher, along with their parents and grandparents, were allowed into the Giants training facility to watch the team practice. Coach John Coughlin welcomed them and then Jackson got to meet some of his favorite players, including Eli Manning, Victor Cruz and Justin Tuck.

He even got to go inside their huddle on the practice field, and was asked to say a few words.

“Let’s crush those Packers!” Jackson shouted, to the delight of the players, who were getting ready to face Green Bay that Sunday. Jackson and his entire family were invited to see Big Blue face off against the Green Bay Packers.

Jackson got one more chance to spend some time with the players when he was invited into the locker room between quarters.

“I feel really good,” Jackson said after meeting the players. He said the day was one of his best ever, “Even better than Christmas.”

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Growing up, Gabriela Carrillo’s three young daughters complained of frequent tummy aches and headaches. The bones in their body, especially their feet, felt like they were burning, they told her.

Carrillo had seen her ex-husband, Luis Zepeda, also suffering from unexplained pain and symptoms.

Then Zepeda’s kidneys failed three years ago, and he went on dialysis. The doctors finally put the puzzle together and diagnosed him with Fabry disease, a rare disease that affects 1 in 40,000 people.

Because it’s hereditary, his family members were tested. Tests revealed his three daughters, Fatima, 13, Kenia, 12, and Gabriela, 8, had inherited the disease and that their 42-year-old dad had inherited it from his mother.

The girls get treatments at Yuma Regional Medical Center every two weeks. The treatments take five hours and consist of intravenous infusions that replace the enzyme.

Although it’s a “very, very long process,” Carrillo is “very, very glad my girls are getting treatment,” she said.

The treatments are expected to become shorter. And for now, the girls travel to Phoenix Children’s Hospital for checkups every six months.

Carrillo works at night as a cocktail waitress at Quechan Casino Resort so she can be free during the daytime to take her girls to doctor appointments and treatments.

To raise awareness of this rare disease, the Carrillo-Zepeda family held a special event March 30 at the Fraternal Order of Eagles. They sold dinner plates with cheese enchiladas, rolled tacos, beans and rice, had a DJ and offered a bake sale and raffles, including a 50/50 (half awarded to the winner and half donated to the Fabry Support and Information Group).

Other prizes included gift cards to various businesses as well as gift baskets and other donated items. All proceeds were donated to FSIG.

Phoenix-based Sarah Cox, a Genzyme patient education liaison, talked about the disease and treatments available. Genzyme is the biotech company that developed the treatment that the Zepeda girls receive.

“I want people to know that although it’s not curable, it’s treatable. You can have a normal, quality life,” Carrillo said. “Fabry testing is free, if anybody thinks they might have it,” she said.
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