SUMMER 2026

INSIDE

30th FSIG ANNIVERSARY

News from the Fabry Support & Information Group

FABRY REFLECTIONS

Earlier diagnosis could have changed my life

By Anthony Waldron
Fabry Patient

The pain started when I was a child—a constant burning in my hands and feet that I couldn't explain.

It would take 36 years before anyone could tell me why.

My name is Anthony, and I was diagnosed with Fabry disease at 43—but my symptoms began when I was about 7.

As a child, I experienced intense burning in my hands and feet, along with heat and cold intolerance and an inability to sweat normally. I also dealt with frequent gastrointestinal issues, often needing to stay close to a restroom at school. Heat made everything worse. While other kids ran and played without thinking about it, I was constantly calculating how to get through the day.

School wasn't just about learning—it was about enduring.

In gym class, I watched other kids run

while I pushed through pain and exhaustion, trying not to fall behind. Sitting in a warm classroom could leave me dizzy and nauseated. I learned early how to tolerate discomfort, but it came at a cost. I stopped raising my hand. I stopped explaining. It was easier to stay quiet than to try to describe something no one else could see.

From the outside, nothing seemed wrong. But every day felt like managing symptoms I didn't understand.

Teachers and coaches questioned my effort. Why couldn't I push harder? Why did I need breaks? Without answers, those questions followed me

See LIFE, page 5

Believe us when we say we're having a bad day

By Narah Cano
FabryDiseaseNews.com

(Feb. 6, 2026) Having Fabry disease means living in an unpredictable body and in a world full of people who often refuse to believe what they can't see.

I have good days when movement feels almost normal, pain sits quietly in the background, and energy allows for work, laughter, and making plans. And then I have bad ones when the burning, tingling, numbness, and intense pain take over without warning. In addition to enduring the physical suffering, the hardest part for me is navigating doubt and judgment when my symptoms fluctuate.

See BAD DAY, page 5

Law enforcement retiree living well on Elfabrio

By Cheisi USA
FabryDiseaseNews.com

(Feb. 20, 2026) Jeff, a retired sergeant with his county sheriff's office, was diagnosed with classic Fabry disease in 2003.

Although he was 33 years old at diagnosis, his symptoms began at a much younger age. At just 8 years old Jeff began to experience what he describes as a burning sensation in his hands and feet—constant discomfort that was both painful and unpredictable. Sometimes this symptom just happened spontaneously; other times it was the result of some type of exertion. It was not until many years later that he

See JEFF, page 3

Finding my tribe at FSIG Get Together

By Susanna VanVickle
FabryDiseaseNews.com

(March 31, 2025) I was met with warm and welcoming smiles when I walked into a boutique hotel near Dallas, Texas, for the recent Fabry Support & Information Group (FSIG) Get Together. There, I spotted a familiar face standing next to a display table. It was Nancy Ngotho, the senior patient education liaison at Sanofi who first educated our family about Fabry disease.

We enjoyed catching up on life updates, including the past several years of my children's Fabry story.

Then, I bumped into Caren Swift on my way to the buffet. She is a highly skilled research nurse who oversaw the administration of the first enzyme replacement therapy that my boys, Michael and Anthony, received in 2019. She has followed my twins' treatment journey ever since, and her unparalleled expertise in Fabry patient care has been an enormous boon for my family.

As we filled our plates with fresh salad, delicious meats, and buttery rolls, we reminisced about my sons' first infusions, their infusions at out-of-state colleges, and their participation in clinical trials.

The luncheon began with an introduction by Lisa Bacon, who is both a Fabry patient and the director of programs at FSIG. She described how the organization was founded and invited us to engage in the community-building opportunities it offers. Next came the guest speaker, Ankit Mehta, a local nephrologist who spoke knowledgeably about kidney health and Fabry disease management. Mehta has played a vital role in my sons' health journeys, overseeing their healthcare for the past several years. Our family is truly fortunate to have him!

After formal presentations from FSIG, Mehta, and other Fabry disease community leaders, we went around the room introducing ourselves and telling snippets of our stories. Seven Fabry disease families were represented at the gathering, and many had never met another person or family with the disease. We delighted in the instant camaraderie of shared experiences.

Fabry patients of all ages, accompanied by their spouses, parents, and children, leaned into the conversations. Some spoke about years of agonizing misdiagnoses, while others discussed the overwhelming burden of consistent symptoms like neuropathy.

In that safe space, we were able to talk about things we usually keep to ourselves.

Columnist Susanna VanVickle (right) and Nancy Ngotho catch up at the FSIG Get Together on March 7, 2026, near Dallas.

Where else can you open up about not knowing what it's like to live without constant diarrhea and having to always know where a restroom is? Where else can you describe the burning sensations in hands and feet, the fever flare-ups, and the ringing of the ears and see everyone nodding along in agreement? It was gloriously refreshing to encounter a world where understanding and solidarity are possible.

Some of the stories were surprising. For example, a woman sitting next to me lost her husband because of Fabry, and all nine of his siblings have the Fabry gene mutation. What are the odds? A toddler with Fabry winced and cried from pain in her feet due to neuropathy. Can symptoms really start that young? Some patients spoke of unconventional remedies that offered symptom relief. How is it that what works for one person with Fabry disease might be useless to another? As questions like these emerged, we received excellent answers from the professionals in the room.

Throughout the meeting, I filled several pages with notes, jotting down health-management strategies, treatment options, resources, other events, and contact information. I left the hotel with stacks of newsletters, booklets, and business cards, and a heart filled with fresh hope. The hours passed quickly, and the meeting left a lasting impact on everyone who attended. I believe each of us felt empowered with knowledge, encouragement, and newfound friendships built on mutual understanding.

After the enriching experience at the Fabry Get Together, I'm excited to share the good news that FSIG exists to help Fabry families and individuals connect. I now have people in my tribe who understand me! I'd encourage you to engage in whatever way you can. Tap into the resources. Explore the wealth of information available about our rare disease. And find a meeting near you or request one—you won't regret it.

Read full story: bit.ly/FabryTribe

JEFF: continued from p. 1

would discover the underlying cause: Classic Fabry disease symptoms appear during childhood or the teenage years and get progressively worse over time.

At the age of 18, Jeff learned he had high blood pressure, which he still deals with today. Despite his health challenges, at age 21 Jeff followed his father into a career in law enforcement. At 22, when his feet and legs started to swell to the point he was having difficulty tying his shoes, Jeff was thought to have Milroy's disease, a form of lymphedema.

In March 2003, Jeff was shown to be in kidney failure, and it was through a kidney biopsy that he finally received his diagnosis of classic Fabry disease. Jeff began dialysis in April of that year—3 days a week, 4 hours each day—and continued for 4 months. He then received a kidney transplant, with his father as the living donor, in August 2003.

Following his transplant recovery, Jeff began enzyme replacement therapy (ERT) for treatment of Fabry disease. On July 3, 2003, Jeff had his first ERT infusion—a treatment that would take place every 2 weeks for 7-8 hours each time.

In 2019, during an emergency room visit for difficulty breathing, Jeff was told he had a third-degree heart block, meaning the upper and lower chambers of his heart were no longer working together. To address this, Jeff received a pacemaker.

In January 2024, Jeff changed his treatment to Elfabrio. Prior to starting treatment, Jeff discussed with his doctor the common side effects such as common cold, headache, diarrhea, and fatigue as well as the potential for severe allergic reactions, including anaphylaxis (see elfabrio.com for more safety information). Almost 2 years into treatment with Elfabrio, Jeff reports that he is tolerating it well.

Jeff is now retired after almost 30 years in law enforcement, and he enjoys staying active by working in a grocery store and fishing in a local creek with his grandsons. Since Jeff's diagnosis, 10 family members have also been tested for and diagnosed with Fabry.

Read full story: bit.ly/JeffsJourney

After three decades, FSIG gets new leader

From FSIG Staff


Johnson


Bacon

For many years, Jack has dedicated his time, passion and heart to serving the Fabry community and helping grow FSIG into the organization it is today.

Through advocacy, education, support programs and countless personal connections, his leadership has touched the lives of so many families across the rare disease community.

As organizations evolve, leadership transitions are a natural part of ensuring continued growth and sustainability. We would like to share that Lisa Bacon has stepped into the role of Executive Director of FSIG.

Lisa brings a strong commitment to the mission of supporting individuals and families affected by Fabry disease. She has already been actively involved in the organization and looks forward to continuing the important work of advocacy, education, awareness and patient support alongside our community, volunteers, partners, and board members.

Jack will continue to support the organization as senior advisor during this transition, and we are deeply grateful for his 30 years of dedication, vision and service to the Fabry community.

Our mission remains unchanged: to provide support, resources and hope to those impacted by Fabry disease while continuing to advocate for awareness, research and access to care.

Thank you for being part of this community and for your continued trust and support as we move forward together.


The full FSIG staff, from left: Connie Baldwin, director of operations; Cheryl Nolte, accounts manager; Jack Johnson, senior advisor; Sabina Kineen, program associate; Barbara Peuster, program associate; Lisa Bacon, executive director.

Insights from Fabry nephrologist at UCLA

By Melissa James
FSIG Contributor

FSIG had the opportunity to interview Dr. Anjay Rastagi from the University of California, Los Angeles, about his work and insights on the future of Fabry.

Q: What is your current title and program?
Professor of Medicine and Clinical Chief of Nephrology at UCLA Health; director of CORE Kidney, which includes the Fabry Program; founder of the Bruin Beans Health Club at UCLA, a premier mentorship program for undergraduates.

Q: What inspired you to pursue a career in healthcare?
My father was a physician in India, and watching him work was very inspiring. From a young age, I was moved by the profound difference he made in people's lives—his care brought hope to patients, comfort to their families and strength to our community. Even before I understood the science of medicine, I felt the deep sense of purpose and compassion that guided him every day. That sense of meaning was powerful, and it sparked a passion in me to follow in his footsteps and to help others.

Q: How does the UCLA CORE Kidney Health program help patients with Fabry disease?
CORE stands for Clinical excellence, Outreach, Research, and Education. All of our patients with Fabry are seen here, which is really important—we're one of the few if not only programs treating Fabry that is led by a nephrologist. We see all Fabry patients irrespective of whether they have renal involvement or not. CORE Kidney is a comprehensive program that covers all aspects of care, including mental health—not just for patients but also for caregivers. Stress and anxiety often stems from fear of the unknown, so we do our best to educate patients about Fabry and connect them with other patients who have it and with specialists who can help treat them.

Q: Can you describe a particularly challenging case that shaped your approach to Fabry?
There is one that stands out. I was giving a presentation about Fabry, and afterward, one of the attending nephrologists approached me and said he thought his niece might have the disease. We tested her, and it came back positive. During her growth, she had been experiencing a lot of anxiety and felt like no one was paying attention to her symptoms. She started treatment, and she now has

two kids and is doing well. Awareness about these rare conditions is important. It is not rare if you have it.

Q: What advancements in Fabry research/treatment are you most excited about?
I feel a genuine sense of excitement about the future of nephrology and the advancements that are transforming the lives of our Fabry patients. Breakthroughs in genetics, immunology and artificial intelligence fill me with hope—we are standing on the edge of a new era in medicine. It's inspiring to witness discoveries that can offer patients longer, healthier and more fulfilling lives. These are truly exhilarating times.

Q. How do you approach conversations about Fabry treatment options with patients?
A shared decision-making approach with patients and their families is critical, but first, they have to know what they're dealing with and what their options are. Education and awareness is very important. Females with Fabry disease presents a different kind of challenge—from the misconception that females are carriers to the spectrum of manifestations based on the inactivation of the x chromosome. Another group that can be challenging is the younger population when they don't have any over manifestations of the disease. Once again, education is the secret sauce.

If you could give Fabry patients one piece of advice, what would it be?
My advice is simple: Be your own best advocate. No one has more stake in the game than themselves. Knowledge is power once it is applied.

LIFE: continued from p. 1

—and eventually, I started to believe them.

When I was told it might be anxiety—that it could all be in my head—I didn't know what to think. Over time, that uncertainty turned inward. I questioned my own limits and whether I was somehow falling short.

For decades, my symptoms were minimized or dismissed—while Fabry disease, as a progressive genetic disorder, continued affecting my body. Living without a diagnosis didn't just impact my physical health—it shaped how I saw myself. It created frustration, self-doubt and a persistent sense of being misunderstood.

What I needed most was simple: for someone to look deeper, sooner. When I was finally diagnosed at 43, I wasn't devastated—I was relieved.

For the first time, everything made sense. There was a name for what I had experienced since childhood. I felt validated. The doubt I had carried for years began to lift. It wasn't in my head. It never was. That moment didn't erase the past, but it reframed it.

Family screening later revealed my case was a de novo mutation—a spontaneous genetic change—with no family history to guide earlier detection. That's why awareness matters. Rare diseases don't always follow clear patterns, and patients shouldn't be dismissed when their symptoms don't fit expectations.

Earlier recognition could have changed the course of my life. Today, I participate in a clinical trial because I don't want others to go through what I did. For me, that means showing up, committing my time, and contributing to research that could help someone else get answers sooner. It's a way to turn years of uncertainty into something meaningful.

Because the emotional toll of waiting for answers is real.

No child should grow up feeling dismissed or misunderstood because their symptoms aren't visible. No patient should spend decades questioning their own reality. And no one should have to carry the weight of being told it's "all in their head" when it isn't.

Rare diseases like Fabry may be uncommon, but their impact is profound. Earlier diagnosis doesn't just change treatment—it gives people their lives back sooner.

BAD DAY: continued from p. 1

On bad days, even a light touch can hurt. Walking feels impossible, and fatigue becomes crushing. It's not the kind of fatigue that sleep fixes, but rather the type that weighs down my body and clouds my mind.

And yet, on good days, someone with Fabry disease may appear capable, functional, and even strong, despite having a bad day just before. This contrast creates a dangerous illusion for others: If we were fine yesterday, how can we be struggling today? Being doubted forces us to constantly justify our own experiences and defend rest as a necessity rather than laziness.

Read full story: bit.ly/FabryBadDay

TREATMENT BRIEFS

Alternative therapy a Fabry breakthrough

(April 7, 2026) A pioneering Canadian clinical trial demonstrated that an experimental single-dose gene therapy is a safe, durable, and highly effective alternative for treating Fabry disease. Unlike standard biweekly enzyme replacement therapy (ERT), this gene therapy uses engineered blood stem cells to allow the patient's own body to produce the missing enzyme.

Read full story: bit.ly/FabBreakthrough

What to know about extended Elfabrio dose

(March 31, 2026) For patients stable on enzyme replacement therapy, extending the infusion interval from every 2 weeks to every 4 weeks may reduce the treatment burden associated with this condition. Fewer infusions can make it easier for patients and caregivers to integrate treatment into their daily lives, which may support long-term adherence and continuity of care.

Read full story: bit.ly/alfa-dose

Long-term efficacy of Migalastat in females?

(Jan. 12, 2026) A recent post hoc analysis of long-term efficacy of migalastat in females with Fabry disease was published in the *Journal of Medical Genetics*. This data highlights the disease severity and burden of females with Fabry disease and supports the long-term efficacy of migalastat. The FACETS (NCT00925301) clinical trial was a double-blind, placebo-controlled study on kidney globo-triaosylceramide in patients with Fabry. *Read full story: bit.ly/MigalastatFemale*

WORLD BRIEFS

INDIA: Funding to run out for 100 rare-disease kids

(Feb. 16, 2026) Some of the country's most vulnerable children reached out to Prime Minister Narendra Modi with a heartfelt appeal for the 'gift of life.' While the National Policy for Rare Diseases has provided critical support, more than 100 children have now exhausted the Rs 50 lakh financial assistance limit.

Read full story: bit.ly/GiftofLifeAppeal

INDIA: Fabry: the LSD misdiagnosed for decades

(May 2, 2026) 28-year-old Arjun spent his entire childhood and teenage years visiting doctors for excruciating burning pain in his hands and feet, mysterious red-purple skin rashes on his torso, inability to sweat properly, and recurring fevers. Understanding Fabry disease is crucial because early enzyme replacement therapy can prevent life-threatening complications, including kidney failure, heart attacks, strokes, and premature death in the 40s-50s; yet diagnosis is often delayed until irreversible organ damage happens.

Read full story: bit.ly/LSDMisdiagnosed

PHILIPPINES: Doctors push for national registry on rare diseases

(Feb. 27, 2026) Health experts said a unified national registry for rare diseases in the Philippines will raise more awareness and support for Filipinos diagnosed with rare conditions. To further support Filipinos diagnosed with such conditions, the Philippine Health Insurance Corp. (PhilHealth) also aims to expand its Z-benefits to ten rare genetic diseases this year.

Read full story: bit.ly/UnifiedRDNR

S KOREA: Kakao, Sanofi partner to harness medical data for AI models

(April 6, 2026) Kakao Healthcare signed an MOU with Sanofi-Aventis Korea to conduct real-world evidence research and develop AI solutions using medical data, The Korea Times reports. The companies said they will run RWE studies using healthcare data and develop AI models through federated learning. Kakao Healthcare will provide platforms and technologies based on real clinical data, while Sanofi will contribute AI, digital healthcare and medical science expertise. The companies said they will explore early Fabry disease diagnosis and tailored asthma management models.

Read full story: bit.ly/SanofiAI

S KOREA: GC Biopharma shows new LSD pipeline

(Feb. 11, 2026) GC Biopharma said Wednesday it presented the current status of its therapeutic development for lysosomal storage disorders at the WORLD Symposium 2026 last week. GC Biopharma also participated in the global phase 1/2 investigator meeting for the Fabry disease treatment HM15421/GC1134A.The company shared safety data, preliminary pharmacokinetic (PK) and pharmacodynamic (PD) data from Cohort 1 subjects, and discussed future clinical strategies.

Read full story: bit.ly/LSDPipeline

Making strides toward greater awareness

FSIG's virtual walk kicks off awareness month

By Susanna VanVickle
FabryDiseaseNews.com

(April 28, 2026) One way to participate in Fabry Disease Awareness Month (April) was by joining a virtual run/walk fundraiser hosted by the Fabry Support & Information Group.

FSIG shared four simple steps for joining the campaign, which I decided to follow. I figure activism doesn't always require a Herculean effort. If enough of us take small steps, we will see a shift in the national, and possibly global, awareness of Fabry disease.

1. **Register for the virtual walk.** Easy.
2. **Order the T-shirt.** Done.
3. **Spread the word.** While social media offers a fast, wide-reaching way to shine a light on Fabry disease, I do not have any form of social media. So I made efforts to weave Fabry awareness into my conversations with friends, neighbors, and colleagues.
4. **Walk.** Since this was my first attempt at rallying people to walk for a cause, and I don't yet have contacts for other Fabry families in my area, I did not go all out in organizing an event. However, I did invite families from my own neighborhood to meet at a nearby park to walk in support of Fabry Awareness Month. A small group gathered on a cloudy weeknight and walked around the park. This casual yet purposeful venture was easy to pull off.

Neighbors gather for a Fabry awareness walk in Irving, Texas

Like a marathon, Fabry disease awareness is a long-distance endeavor, and each step in the right direction gets us incrementally closer to our goal. Staying active in the Fabry community year-round is an important way to keep the momentum going.

Read full story: bit.ly/FabryAware

Genomenon, Amicus partner to advance Fabry Disease

Press Release

(April 2, 2026) Genomenon today announced a strategic sponsorship from Amicus Therapeutics to accelerate awareness, diagnosis, and research for Fabry disease.

Through this collaboration, Genomenon produced literature-derived real-world evidence by applying fit-for-purpose AI and expert review to curate and classify variants in the GLA gene. The resulting variant evidence has been made freely available to the global clinical and research community through the Mastermind Genomic Intelligence Platform and submitted to ClinVar, the widely used public archive of human genetic variants hosted by the National Center for Biotechnology Information.

By broadening access to curated GLA variant evidence in both Mastermind and ClinVar, Genomenon and Amicus aim to support faster, more confident interpretation of genetic tests and reduce delays to diagnosis for patients and families affected by Fabry disease.

Read full story: bit.ly/genomenon

Fabry Awareness Month focuses on community, strength, support

By Marisa Wexler, MS
FabryDiseaseNews.com

(April 2, 2026) April is Fabry Awareness Month, and advocates around the world are working to uplift and connect the community, increase awareness about Fabry disease, and support fundraising efforts.

For this year's awareness campaign, the Fabry International Network (FIN) has selected the theme: "Living with Fabry, Your Unique Strength." According to FIN's website, this theme was selected because "Fabry is not only a diagnosis or a list of symptoms. It's a life. A rhythm. A constant process of adapting, advocating, coping, resting, showing up, and starting again. And within that lived experience, there is often strength not always loud or obvious, but real."

During awareness month, the MPS Society encourages people to get involved and support the Fabry community. "Our members face so many challenges because of their symptoms and we are here to make life as easy as possible so they can live life to their full potential," according to the society's website.

Read full story: bit.ly/FabryAM

FSIG

FSIG is a support group dedicated to dispensing information and encouraging mutual self-help as a means of emotional support.

FSIG was formed in 1996 by two Fabry patients and supportive family members with the hope that their particular understanding of this disease, combined with experience gathering information and working with doctors could benefit others.

FSIG is a nonprofit, tax-exempt organization and relies on charitable contributions to provide services to those with Fabry disease, their families and supportive others. Donations may be sent to the address below.

Please feel free to make copies of the FSIG Newsletter to share with your family, friends and others. We encourage anyone interested in FSIG or the newsletter to contact us so we can make sure you receive the next issue.

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GENE THERAPY

AMT-191 shows promise, but paused over safety

By Marisa Wexler, MS
FabryDiseaseNews.com

(Feb. 12 2026) The experimental gene therapy AMT-191 appears to be working as intended in an early clinical trial, developer uniQure said, but the company is pausing part of the study pending further investigation into severe liver damage cases.

AMT-191 is designed to deliver a healthy version of the gene encoding alpha-Gal A to cells, allowing the body's cells to produce a functional version of the enzyme that can clear toxic fatty acid deposits. uniQure is sponsoring a Phase 1/2 clinical trial to test the gene therapy in a dozen men with Fabry disease, ages 18 to 50, who have had an inadequate response to ERT.

Read full story: bit.ly/AMT-191Pause

Sangamo seeks accelerated approval of gene therapy

By Michela Luciano, PhD
FabryDiseaseNews.com

(March 12, 2026) Sangamo Therapeutics said it advanced the rolling submission of a biologics license application (BLA) seeking U.S. accelerated approval of its gene therapy candidate isaralgagene civaparvovec (formerly ST-920) for Fabry disease, following positive data from a clinical trial.

Accelerated, or conditional, approval allows experimental therapies to reach the market based on early evidence suggesting they are likely to benefit patients. Full approval from the U.S. Food and Drug Administration (FDA) depends on additional clinical trial data confirming the treatment's benefit.

The company is seeking approval through a rolling review process, which allows sections of a BLA to be filed as they are completed rather than waiting until the entire dossier is finalized. Sangamo, which started the application process earlier this year, announced in a company press release that it has now submitted the preclinical and clinical modules of the application to the FDA for review.

A companion diagnostic test has also been submitted to the FDA's Center for Devices and Radiological Health and accepted for review through the agency's Premarket Approval pathway, the company said. If approved, the test would be used to screen patients before treatment to determine whether they are suitable candidates for the therapy.

Read full story: bit.ly/SangamoGT

Gene therapy trial starts in Alabama

The University of Alabama at Birmingham is taking part in a groundbreaking Phase I/II clinical trial testing a one-time, intravenous investigational gene therapy that could transform care for patients with classic Fabry disease.

Read full story: bit.ly/FabryUAB

CELL THERAPY

GT-GLA-S03 gets orphan designation from US FDA

By Marisa Wexler, MS
FabryDiseaseNews.com

(March 19, 2026) The U.S. Food and Drug Administration has granted orphan drug designation to GT-GLA-S03, an experimental cell therapy being developed by Glafabra Therapeutics for Fabry disease.

The FDA gives this designation to investigational therapies for diseases that affect fewer than 200,000 people in the U.S. It offers incentives such as tax credits and fee waivers, as well as seven years of market exclusivity. This encourages companies to develop treatments for rare diseases.

Read full story: bit.ly/OrphanGT-GLA-S03

GALAFOLD

Brain health remains stable in Fabry patients

By Marisa Wexler, MS
FabryDiseaseNews.com

(Jan 22, 2026) Markers of disease-related tissue damage in the brains of Fabry disease patients treated with Galafold (migalastat) tend to remain stable over time, a study found.

Fabry patients with high blood pressure may be more likely to have damage in the brain, according to the study, suggesting that treatments to control blood pressure may help prevent disease-related brain damage. This observational FAMOUS trial was funded by Amicus Therapeutics, the company that sells Galafold.

Read full story: bit.ly/GalafoldBrain

LUCERASTAT

Long-term use of lucerastat may protect kidneys

By Margarida Maia, PhD
FabryDiseaseNews.com

(Jan. 29, 2026) Long-term dosing with lucerastat, an experimental substrate reduction treatment from Idorsia, may slow the loss of kidney function in adults with Fabry disease, perhaps by reducing the buildup of toxic fatty molecules in the body's cells, a global clinical trial has found.

The oral therapy—given to some study participants for more than six years—was generally safe and well tolerated, Idorsia noted in a company press release announcing the results of the Phase 3 study, dubbed MODIFY (NCT03425539), and its open-label extension (NCT03737214). The company noted what it called a "promising effect" seen with lucerastat.

Read full story: bit.ly/luceratrial

Phase 3 program planned for lucerastat

By Marisa Wexler, MS
FabryDiseaseNews.com

(Feb. 26, 2026) Following discussions with regulatory authorities in the U.S. and Europe, Idorsia has outlined a new Phase 3 registration program to evaluate its experimental oral therapy, lucerastat, for its effects on kidney outcomes in people with Fabry disease.

If the studies are successful, Idorsia said the data could serve as the basis for a potential regulatory submission for lucerastat. The company said a filing could occur as early as 2029.

Lucerastat is an experimental substrate-reduction therapy. This type of treatment aims to reduce the buildup of Gb3 by blocking its production. Because its mechanism does not depend on alpha Gal-A activity, lucerastat is designed to be suitable for patients regardless of their underlying mutation type. Idorsia is developing it as a potential first oral treatment option for all Fabry patients.

Idorsia previously sponsored a Phase 3 study, MODIFY (NCT03425539), which tested twice-daily oral lucerastat versus placebo in more than 100 adults with Fabry disease. The study did not meet its main goal of showing that lucerastat reduced neuropathic pain (pain caused by nerve damage) compared with placebo.

Although lucerastat did not demonstrate a benefit for pain relief, data from MODIFY and its open-label extension (NCT03737214) showed reductions in Gb3 levels. Long-term analyses also suggested a slower decline in kidney function compared with patients' prior historical trajectories.

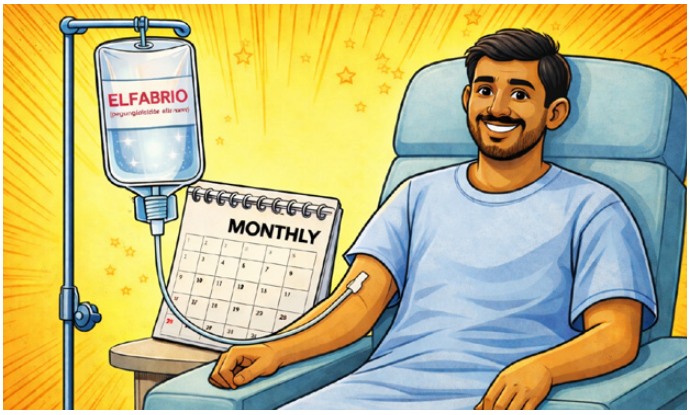
Idorsia plans to run two new Phase 3 trials to evaluate lucerastat's effects on kidney function. Idorsia did not provide a timeline for when the two studies are expected to begin enrolling patients.

Read full story: bit.ly/IdorsiaP3

ELFABRIO

Monthly Elfabrio dosing approved in EU

By Michela Luciano, PhD
FabryDiseaseNews.com



(March 26, 2026) A monthly dosing regimen for the infusion therapy Elfabrio (pegunigalsidase alfa) has been approved in the European Union for adults with Fabry disease who are stable on enzyme replacement therapy.

Under the newly cleared regimen, approved by the European Commission, eligible patients may now receive the ERT every four weeks at a dose of 2 mg/kg instead of every-two-week dosing at 1 mg/kg. For those with stable Fabry, this treatment schedule is expected to provide a more convenient dosing alternative.

According to developers Chiesi Global Rare Diseases and Protalix Biotherapeutics, the decision follows a positive opinion from the Committee for Medicinal Products for Human Use, a branch of the European Medicines Agency responsible for evaluating new drugs and recommending whether they should be approved in the EU.

Importantly, the older regimen continues to be the only option for patients in the EU who are not stable on ERT. It also remains the only approved dosing regimen in the U.S.

Read full story: bit.ly/elfabrioEU

5-year stability with monthly dose of Elfabrio

By Margarida Maia, PhD
FabryDiseaseNews.com

(April 23, 2026) Elfabrio, when given every four weeks, was generally safe and helped most Fabry disease patients maintain stability for up to five years. The therapy was particularly effective for women and those who did not develop antibodies against the replacement enzyme.

That's according to interim results of the ongoing BRIGHT F51 open-label extension study testing the long-term efficacy and safety of Elfabrio at 2 mg/kg of body weight every four weeks in 29 adults with Fabry who had already received the monthly ERT for 1 year.

Read full story: bit.ly/Elfabrio5

STUDY SYNOPSES

Eye abnormalities may signal heart disease in Fabry

(Feb. 19, 2026) People with Fabry disease have abnormalities in the blood vessels within the eye, and these abnormalities may be indicative of heart disease in Fabry patients, a first-of-its-kind study reports.

Researchers also found an association between eye vessel abnormalities and more inflammation in people with certain Fabry-causing mutations that are associated with heart disease. The scientists said this finding suggests anti-inflammatory therapies may be beneficial in these patients, though further research is needed.

Read full story: bit.ly/EyeAbHeart

Gene mutation, kidney health linked to lower stroke risk

(Jan. 12, 2026) Nearly one in five adults with Fabry disease experienced a stroke over roughly a decade of follow-up, with risk shaped by genetic factors, kidney function, and coexisting autoimmune disease, a large U.K. study found.

Preserved kidney function and carrying the disease-causing N215S mutation—which is often linked to a late-onset and generally milder form of Fabry—were both associated with a lower risk of stroke, by up to 60%. In contrast, having a coexisting autoimmune disease raised stroke risk more than threefold.

Read full story: bit.ly/GeneKidney

Fabry newborn screening test misses girls

(April 16, 2026) Since its implementation in 2017, newborn screening (NBS) for Fabry disease in Tennessee has sharply increased early diagnosis for baby boys, but continues to miss affected girls, a study found.

From 2017 to 2024, statewide screening identified 25 boys with Fabry but no girls. Yet clinical data from roughly the same period showed the condition affected boys and girls in nearly equal numbers.

Read full story: bit.ly/Fabrytestgirls

Fabry, aHUS & heart disorder an 'unusual trilogy'

(March 10, 2026) A teenage boy who was diagnosed in childhood with atypical hemolytic uremic syndrome (aHUS) was later found to also have Fabry disease and an inherited heart condition called hypertrophic cardiomyopathy—what researchers described as an "unusual trilogy" of rare diseases.

The two additional genetic conditions were identified only after new symptoms appeared during adolescence, including worsening kidney function and increasing thickening of the heart muscle. Additional testing, including genetic analysis, eventually confirmed both diagnoses.

Although targeted treatment was started, the boy's kidney function continued to decline, and the thickening of his heart muscle did not improve. The researchers suggested this may reflect organ damage that had already progressed by the time treatment began, along with the combined effects of multiple genetic conditions.

Read full story: bit.ly/FabryHDaHUS

Fabry symptoms often mimic IBS in adult patients

(March 5, 2026) Routine screening for gastrointestinal (GI) issues should be a standard part of clinical care for Fabry disease, according to a new study that found more than 70% of patients experience GI symptoms.

The research highlights that roughly half of adults with the genetic disorder experience moderate to severe digestive problems, which are directly linked to a lower quality of life and increased physical pain.

Because these issues are so closely tied to overall disease severity, researchers are urging doctors to implement "routine screening for gastrointestinal symptoms at diagnosis and annually thereafter."

Read full story: bit.ly/IBSmimic

Children wait years for Fabry diagnosis in China despite symptoms

(April 9, 2026) Children with Fabry disease typically experience long delays before receiving a diagnosis, despite symptoms affecting multiple organs that often emerge in early childhood, a nationwide study in China has found.

Overall, a median diagnostic delay of at least four years was seen among the more than 60 children in the Asian nation who were involved in the new study.

The findings also showed that boys tended to develop Fabry symptoms earlier and experience more severe disease, with nerve-related pain emerging as the most common early symptom. Both boys and girls also experienced absent or reduced sweating, heart rhythm abnormalities, and eye issues, the data showed. Some boys also had signs of kidney involvement.

Read full story: bit.ly/chinafabrykids

PREDICTORS OF CARDIOVASCULAR EVENTS IN FABRY

Read the results of this important study:

bit.ly/FabryPredictors

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