SPRING 2026

INSIDE

30th FSIG ANNIVERSARY

News from the Fabry Support & Information Group

CELEBRATING THREE DECADES OF ADVOCACY

From kitchen table to global advocate

Celebrating FSIG's journey

Visiting North Dakota Sen. Kevin Cramer's office in 2019

By Jack Johnson
FSIG Executive Director

What started around a kitchen table in 1996 has grown into something truly special. The Fabry Support & Information Group (FSIG) began with a simple but powerful mission: make sure no one faces Fabry disease alone. Three decades later, that promise still guides everything we do.

How We Got Started

When Jack, Debra, and Kathy Johnson first gathered to create FSIG—encouraged by Dr. Robert Desnick—they knew the Fabry community needed a voice. In 1997, we published our very first newsletter (just 100 copies!) and launched the first website dedicated to Fabry patients. Those modest beginnings planted seeds that would grow into a worldwide network of support, education, and advocacy.

The early 2000s brought major milestones. Along with you we wrote letters to the FDA, attended meetings, and helped influence the approval of enzyme replacement therapy in 2003. By 2005, we participated in the formation of Fabry International Network, connecting patients and advocates across

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Q&A with Fabry geneticist Dr. Robert Hopkin

For this edition of our Physician Spotlight, we are honored to feature Dr. Robert Hopkin, a professor with the division of human genetics at Cincinnati Children's Hospital Medical Center. He sees both pediatric and adult patients.

Q: What inspired you to pursue a career in healthcare? I originally wanted to be a field biologist but realized people weren't going to pay me to do that, so I went to medical school. I knew I didn't want to be a surgeon or an internal medicine specialist. I was leaning toward pediatrics, but I

learned a lot about rare diseases, so I went into genetics.

Q: What drew you to Fabry? I got into genetics to take care of kids with birth defects and disabilities. One day, my boss came in and said he needed someone who was passionate about Fabry disease. I didn't know anything about it. When he told me more about it, I became intrigued and said, "What do I have to do?" This was in 1997, when there was no treatment and it was seen primarily as an adult male disease.

Q: Can you describe any challenges that shaped your approach to Fabry?
I met a lot of women and children who had symptoms despite what we thought about the disease, and that motivated me. You couldn't see [their] pain, but they would tell me they were in pain. There was a 16-year-old boy who diagnosed

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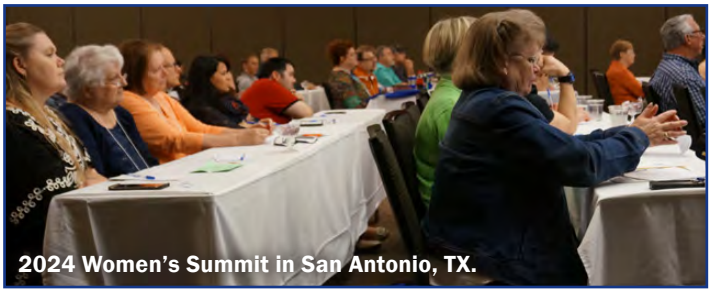
2024 FSIG Conference group photo - From the FEFC in La Jolla, CA.


FSIG Executive Director Jack Johnson with renowned Fabry specialist Dr Robert Desnick in 2002.

Entry to 2016 FSIG Expert Fabry Conference.


2024 Women's Summit in San Antonio, TX.

the globe. Every step forward was guided by one question: What does our community need?

Changing the Conversation

One of our proudest achievements has been advocating for women and girls with Fabry disease. For too long, females were told their symptoms weren't from Fabry or didn't need treatment. We refused to accept that. By partnering with researchers, clinicians, and industry leaders, we've helped change the medical conversation. Today, more women are being diagnosed, validated and treated with the care they deserve.

We've also been champions for the youngest members of our community. By partnering with "Testing for Tots" and our ongoing push for newborn screening, we're working to ensure babies get diagnosed early—sparing families years of uncertainty and giving children the best possible start.

Support That Shows Up

FSIG isn't just about advocacy—we're here in practical ways too. Since 2011, our Fabry Assist program has provided over 750 financial awards to families who needed help. Whether it's covering travel to appointments or easing other burdens, we show up when it matters most.

We've also earned a seat at important tables. From the National Kidney Foundation to international medical conferences, FSIG represents your voice in conversations about research, treatment, and policy. We've advocated on Capitol Hill, collaborated on patient-focused reports, and joined coalitions to amplify the rare disease community.

What's Next

As we look ahead, our focus remains clear: expand newborn screening so more babies are identified early, support newly diagnosed families from day one, and keep fighting for care that honors every person's Fabry experience—regardless of age, gender, race, or how the disease shows up for them.

We're grateful to be part of this incredible community and the broader rare disease family. Being surrounded by partners who listen, collaborate and truly put patients first reminds us that real progress happens when we move forward together. From those first 100 newsletters to a global network of support, FSIG's story is really your story. Thank you for letting us walk alongside you, today and always.

Want to learn more about FSIG programs or get involved? Visit our website or reach out—we're here for you.


FSIG annual Fun Run/Walk in 2014.

Eye scan spots nerve damage

By Steve Bryson, PhD
FabryDiseaseNews.com

(Oct. 16, 2025) Noninvasive in vivo confocal microscopy (IVCM) detected signs of nerve damage in the cornea—the eye's transparent outer layer—in people with Fabry disease, who also had higher levels of inflammatory immune cells in the cornea than healthy individuals, a study found.

"IVCM provides parameters that reliably indicate corneal nerve damage and inflammatory activation in patients with [Fabry disease]," the researchers wrote.

The study, "Corneal neuro-immune crosstalk in Fabry disease: An in vivo confocal microscopic study," was published in the *Journal of Neuroimmunology*.

Gb3 can accumulate in the densely packed small nerve fibers within the cornea, which convey sensory information about touch, pain, and temperature. Damage to these small nerve fibers—a condition known as corneal small fiber neuropathy—can cause severe eye pain and other serious complications.

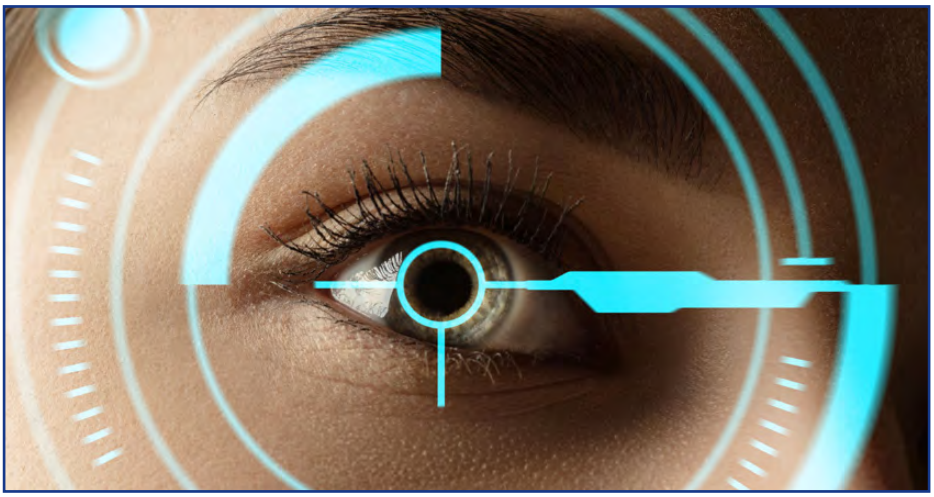
IVCM allows for direct visualization of the small nerve fibers in the cornea. It can also detect immune cells, primarily corneal Langerhans cells, which act as local sentinels of the immune system and can promote an inflammatory response upon the detection of tissue damage.

While IVCM has been used to detect Gb3 deposits in the cells of the cornea, studies focusing on corneal nerve changes in Fabry are limited, with no previous studies investigating corneal inflammatory cells.

"In-depth understanding of corneal nerve changes and innovated evaluation of corneal inflammatory status in patients with [Fabry] are urgently needed for the further application of IVCM as a valuable marker of [Fabry]," wrote the research team in China, who applied IVCM to both eyes of 31 Fabry patients (13 females and 18 males), ages 14-64, and those of 25 age- and sex-matched healthy individuals, who served as a control group.

Fabry patients also had more nerve tortuosity (or abnormally twisted nerve fibers) in the cornea, a sign of nerve damage..

Read full story: bit.ly/Fabry-Eye



FABRY TESTING

Troponin blood tests reliably rule out Fabry

By Lindsey Shapiro, PhD
FabryDiseaseNews.com

(Sept. 18, 2025) High-sensitivity blood tests to measure troponin — a biomarker of heart cell damage — can be used to rule out significant cardiomyopathy, a heart condition, in people with Fabry disease, according to a recent report.

In clinical settings, this could help physicians promptly identify patients in need of more substantial monitoring and treatment, while avoiding resource-intensive MRI imaging for people at low risk.

"Taken together, serial [high sensitivity troponin] assessment appears to be a reasonable approach for detecting early myocardial [heart muscle] involvement in patients with genetically confirmed [Fabry disease]," researchers wrote.

The study, "The Utility of High-Sensitivity Troponin to Detect Cardiomyopathy in Patients With Fabry Disease," was published in *JIMD Reports*.

"The identification of a readily available blood biomarker that could reliably exclude significant FC [Fabry cardiomyopathy] would allow us to prioritise cardiac MRI for patients for whom it is most likely to impact their care," the researchers wrote.

To date, blood biomarkers have served a supportive role in monitoring Fabry's heart disease.

Read full story: bit.ly/FabryTest

Echocardiograms could detect early heart disease in Fabry

By Steve Bryson, PhD
FabryDiseaseNews.com

(Nov. 20, 2025) Noninvasive echocardiogram imaging may serve as an early marker for left ventricular hypertrophy (LVH), a disease of the left heart muscle, in adults with Fabry disease, a study suggests.

Imaging measurements could reliably distinguish Fabry patients among individuals with LVH, with Fabry men, in particular, showing markedly worse heart function than LVH patients without Fabry, the data showed.

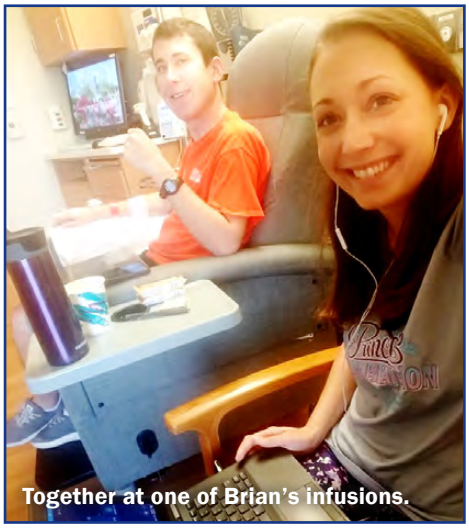
"This is a clinically relevant distinction and has implications for the imaging assessment of patients with LVH," researchers wrote in the study, "Differentiating Anderson-Fabry Disease from Other Causes of Left Ventricular Hypertrophy: Novel Insights from Left Atrial Strain Imaging."

Read full story: bit.ly/FabryEcho

Finding our new normal & truly living

By Tia Jones
Co-Founder of Testing for Tots

Living with Fabry... those weren't words I could even comprehend in 2017. I was planning our wedding. My fiancé, Brian, was interviewing for his medical residency program. I was interviewing for jobs. I flew home from an interview to find my fiancé sitting on the couch in the dark. He was in tears. He told me he had been diagnosed with Fabry disease.


Together at one of Brian's infusions.

The next two months went by in a blur. Genetic testing. Genetic counselor. Mapping out our family tree. Packing to move. Driving 18 hours across the country to Minnesota. That summer felt like we were trying to get our bearings in a new place, new jobs, new treatment, new everything.

Eventually, a new normal came. We'd pack up dinners and walk to the clinic for infusions, and Brian and I started wondering why only four states did not allow copay assistance. While we started digging into Minnesota state law during infusions, I could not help but wonder, what if? What if Brian had been diagnosed earlier? What if he could have gotten treatment before his kidneys were halfway scarred?

What other families are walking around with numb feet, upset stomachs and losing their hearing, while having silent damage done to their kidneys, heart and brain? What standardized testing was available? Our infusion nights became working and brainstorming sessions.

A few months later, we were introduced to Jack Johnson at FSIG, and our fundraiser Testing for Tots was born. We had a successful first year in 2019, and just like anyone else, were reeling on what to do in 2020. We went virtual, and later that fall, lost our first aunt with Fabry disease. We had already set our 2021 date, but candidly, wondered if we should just cherish our time together instead.

Our latest relocation brought us to North Carolina. Of all the moving pieces, the one that most impacted our daily lives was transitioning to home infusions. We've continued our normal of trying to move the needle in different states for newborn screening. One of the most special developments has been sharing Testing for Tots with our daughter, who joined us for our 6th Annual Tasting for Tots.

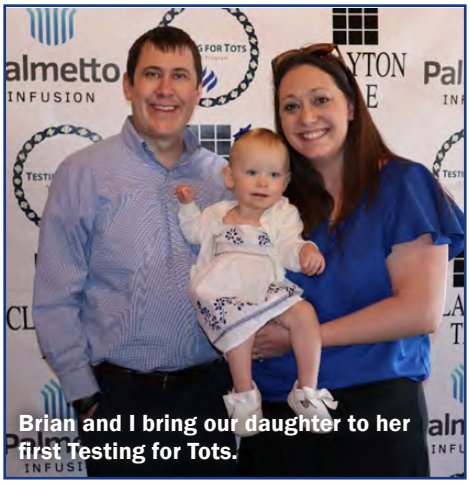
Fast forward nine years. We've found what I yearned for in the early days—but wasn't sure would really come—"finding our new normal." Today we're "living" with Fabry. We've modified our sodium diet so much, I don't even think about what to cook or how to cook—it's what we eat. Meds come in the mail once a month, and I know how to quickly sort through the box to find what goes in the fridge versus the closet. Our nurse rings the doorbell, and I know Nancy by name. Our mission has deepened with Testing for Tots, growing more roots in more states. I think the key for me is not just how we are living with Fabry, but how can we help others to start truly LIVING with Fabry.

Tasting for Tots
Saturday, March 28
4-7 PM
Kennington Family Winery

Please join us in our 7th Annual Tasting for Tots, at this lovely winery between Greenville, SC, and Asheville, NC.. All funds go to advancing newborn screening of Fabry disease and supporting families after diagnosis.

Tickets available until March 20:
fabry.org/testing-for-tots


Our first A Tasting for Testing for Tots fundraiser in 2019, with five of our six Fabry family members.



Brian and I bring our daughter to her first Testing for Tots.

Love can lighten someone's load

By Susanna VanVickle
FabryDiseaseNews.com

(Sept. 30, 2025) Racing down a hill I travel daily, not realizing that my car was going almost as fast as my mind, I caught sight of police lights in my rearview mirror. What?! Not a ticket! Could my day get any worse?

That was the day we were told that my 9-year-old "sweet pea" had the Fabry disease gene mutation. I was driving the short distance from my house to the church—my safe place—where I could unload my burdens and lay bare my heart. As I went, my mind was exploding with questions and emotions. I knew that Fabry had been a possibility, but I'd hoped and prayed Sweet Pea would be spared.

Earlier that summer of 2019, my family's world was rocked with the news that my teenage twin sons, Michael and Anthony, had Fabry disease. I can reflect on those first months of our Fabry journey and remember many ups and downs. Some days, I was the strong mama, researching, pep-talking, smiling, and giving hope. But other days, the long hours of appointments with specialists, genetic counseling, and conversations with medical insurance providers left me feeling weak and spent. The day the cop pulled me over for speeding, I was weak. After coming to terms with the fact that my two sons had inherited a lifelong disease from me, the next hurdle was getting genetic testing for our family members. Thankfully, our first round of tests proved that I was the first in my family line to have the disease. This meant that my sisters and their large families had been spared through a rare version of mosaicism. The second step was testing my other three children.

RELATED STORY

Dad diagnosed with Fabry after dismissing aches and pains as 'aging'

bit.ly/DadShock

We hoped for good news as we waited for the kids' results. Michael and Anthony had been experiencing manifold symptoms before they were diagnosed, and none of my other kids had symptoms at all. So it seemed reasonable to assume they were not recipients of the mutated gene.

The bad news about my little girl was an unwanted surprise. It felt like ripping off a scab and reopening a wound. So there I was speeding to church, and I got caught. The young officer approached my window, and keeping my composure, I politely answered his questions and admitted I hadn't been watching my speed. As he returned to his vehicle to print the ticket, I couldn't hold the tears back.

I knew I should've been paying attention, but I was consumed with thoughts of another Fabry diagnosis for someone I fiercely loved. Now I was having insult added to injury with a heavy fine on top of my heavy heart.

When the officer came to the window holding my ticket, he saw my tear-soaked face and asked if I was OK. "It's not your fault," I whimpered. "I just found out that my little girl has a rare disease." At that point, he softened, asked a few more questions, and said, "I'm not giving you this ticket, and I am sorry for your little girl. I'll keep your family in my prayers."

Today is National Love People Day, which prompted me to tell this story of a stranger who seized an opportunity to lift someone else up. He could've just done his job and been on his way, but instead, he showed me kindness that I won't forget.

Let's go out and love people today in a way they won't forget.

Read full story:
bit.ly/FabryLighten

Help Fabry kids combat depression

By Susanna VanVickle
FabryDiseaseNews.com

(Oct. 28, 2025) We were excited the first time our patient education liaison from pharmaceutical company Sanofi offered to take us out to a fancy restaurant. Several members of my family had just been diagnosed with Fabry disease, and we planned to talk about the condition over dinner. My kids were loving this perk of having a rare disease.

But what was lost on the young ones wasn't so lost on my eldest sons, my husband, and me. The chronic symptoms and lifelong difficulties caused by our family's gene mutation were daunting. I had never heard of some of these things. Neuropathy? Corneal whorls? Angiokeratomas? Anhidrosis?

My kids with Fabry bear the weight of an unpredictable future, chronic relentless pain, tiresome treatment experiments, sleep deprivation, ringing in the ears, heat intolerance, and more.

Author Terry Wardle recommends that, while giving ourselves permission to feel the raw pain and intense distress of that moment, we visit the person we were then, bringing love, acceptance, and hope that we have gained over time.

I believe the depression and dejection that my children suffer can be transformed by that same loving balm. How so? I can revisit painful times in my past with healing grace, and I can teach this technique to my family.

We can permit ourselves to grieve the awful news all over again. Then, we can go a step further and speak to that version of ourselves that was traumatically affected. We can offer hope, acceptance, and grace.

Read full story:
bit.ly/FabryKids

WORLD BRIEFS

CHINA: Insurance gives rare disease patients new hope

(Dec. 15, 2025) Yang Qian, 23, lives with Fabry disease. In 2020, the drug Replagal became available in China, costing around $1,700 per box. By 2021, it was included in China's national medical insurance catalog—cutting the price to $430 per box. This video follows Yang to see how China has built the world's largest health safety net, ensuring no one is left behind.

Watch the video story: bit.ly/RarePath

CHINA: Woman diagnosed with both Fabry and blood disorder

(Nov. 13, 2025) In what researchers say is the first reported case of its kind, a Chinese woman in her 60s was diagnosed with two co-occurring rare conditions: Fabry disease and myelodysplastic syndrome, a disorder in which a person's bone marrow doesn't make enough healthy blood cells. The authors noted that several other family members showed signs of both diseases, with relatives—including the woman's son—carrying the same Fabry-causing genetic mutation alongside low levels of platelets, the cell fragments that help with blood clotting.

Read full story: bit.ly/FabryChina

INDIA: Treatment crisis afflicts rare disease patients

(Dec. 11, 2025) The Indian Medical Parliamentarians' Forum (IMPF), a group of 45 members of parliament who are also medical professionals, has raised an urgent alarm over life-threatening treatment disruptions faced by children and adults living with lysosomal storage disorders. According to data presented in the presentation, around 60 patients have already crossed the existing Rs 50-lakh annual treatment cap, leaving them without any viable means to continue therapy.

Read full story: bit.ly/RareCrisis

CANADA: Study finds stroke risk for young adults

(Dec. 11, 2025) Young adults with Fabry disease in Canada —especially men under 40— are at a significantly higher risk of experiencing a first-time stroke or transient ischemic attack (temporary blockages sometimes referred to as "mini-strokes") than the general population, a new study reveals. The prescribing rates of low-dose aspirin to prevent stroke in Fabry patients have only slightly declined, even though multiple clinical trials have recently shown no benefits.

Read full story: bit.ly/FabryStroke

SPAIN: Progress of Fabry tied to early inflammation

(Oct. 2, 2025) Inflammation, believed to be a secondary or late complication, may instead be an early and active driver of Fabry disease, according to data from a small study in Spain. Researchers found ongoing immune activation and signs of inflammation in all patients, even in those with little buildup of the fatty molecules thought to be the main drivers of Fabry symptoms and organ damage.

Read full story: bit.ly/FabryProg

MAYLASIA: One patient's journey of journey of pain, treadment & hope

(Nov. 19, 2025) Merpati Ahmad may appear ordinary at the surface, but this schoolteacher experiences burning cramps in the muscles of his arms and legs, which become worse in hot weather and after an exercise. Lurking under the surface is the risk of organ damage, especially kidney impairment and heart problems. Merpati Ahmad has Fabry disease.

Read full story: bit.ly/FabryMalaysia

HOPKIN: continued from p. 1

himself after experiencing a lot of pain, abdominal discomfort and fatigue, and no one would listen to him. This was shortly before the approval of Fabrazyme in 2003, and he became one of our first patients to be treated for Fabry.

Q: What advancements in Fabry research/ treatment are you excited about?

There's a lot going on right now. In 2003, there was only one treatment, but now there are currently three FDA-approved treatments. One is a pill and the other two are enzyme infusions. It's exciting to have multiple options. It is going to make life easier for patients.

Q: How do you approach conversations about treatment options?

I update patients on all the treatments available, even the ones they aren't taking. The more they know about the disease, the more they are likely to make good decisions and take good care of themselves.

Q: Why is there a need for more young doctors to get involved in genetic disorder programs?

When I first started as a geneticist, there were no treatable diseases, but that has changed dramatically. As treatments become available, we need people to help care for the patients. If we make it hard for them, they'll miss out on treatment, and that will lead to unnecessary suffering.

Q: What one piece of advice do you have for Fabry patients and their families?

Engage in the discussion and treatment of your disease! They need to know what they are facing and what the options are—and the best way to do that is to be a part of the discussion.

ST-920 boosts kidney, heart function in trial

By Steve Bryson, PhD
FabryDiseaseNews.com

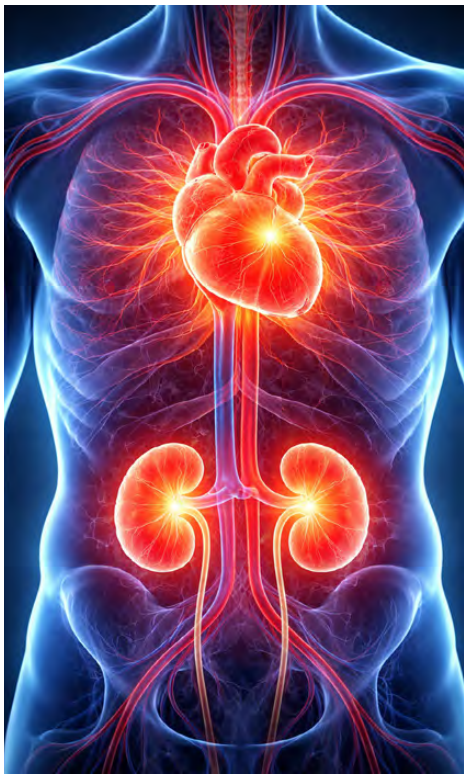
(Dec. 4, 2025) A one-time infusion of gene therapy ST-920 (isaralgagene civaparvovec) improved kidney function and stabilized heart function in adults with Fabry disease for up to two years after treatment, according to new data from a Phase 1/2 clinical trial.

Kidney functional improvements in the STAAR study (NCT04046224) were observed regardless of sex, previous use of enzyme replacement therapy (ERT), type of Fabry disease, or degree of kidney impairment before gene therapy, the data showed.

Sangamo Therapeutics, the therapy's developer, said it met with the U.S. Food and Drug Administration (FDA) and agreed on a proposed efficacy and safety data package to support an approval pathway for the treatment.

The data were presented recently at the International Congress of Inborn Errors of Metabolism 2025 (ICIEM2025) in Kyoto, Japan. Administered by a one-time infusion into the bloodstream, ST-920 works by delivering a healthy copy of the GLA gene to liver cells. It's expected to boost production of a working alpha-Gal A, which can break down and clear fatty molecules from cells, thereby preventing organ damage and easing Fabry symptoms.

STAAR enrolled men and women, ages 18 and older, with Fabry who were previously treated with ERT, had been off ERT for at least six months, or had never received ERT.



Top-line data reported last June showed that among the 32 Fabry patients treated with ST-920, there was an increase in eGFR rate, or an improvement in kidney function, one year after the infusion. The results were similar to those of 19 patients who had been followed for two years.

All 18 patients who began the study on ERT had stopped and remained off ERT after ST-920 treatment. Their blood levels of lyso-Gb3, a marker of Fabry disease, remained generally stable thereafter.

Researchers also noted improvements in measures of disease severity, gastrointestinal symptoms, and quality of life.

The newly reported data showed that patients saw ST-920-induced improvements in the mean annualized eGFR, regardless of sex, previous ERT status, Fabry disease type, and pre-gene therapy eGFR values.

Heart function remained stable for at least one year, as assessed by various imaging measures, including left ventricular mass, the size of the heart's pumping chamber, left ventricular strain, and blood volumes in the heart before and after it contracts.

Before ST-920, 10 participants had measurable levels of antibodies against alpha-Gal A associated with previous ERT use, which can limit the treatment's efficacy. After ST-920, antibody levels decreased markedly in nine patients and eventually became undetectable in eight.

Sangamo also reported on a Fabry patient who had withdrawn from ERT during the study, but restarted ERT as directed by a physician. The patient received ST-920 more than two and a half years ago, and maintained higher-than-normal levels of alpha-Gal A activity and stable levels of lyso-Gb3.

The most common adverse events reported during STAAR were fever, COVID-19, the common cold, and headache, all of which were resolved with standard medical treatments.

Read full story: bit.ly/FabryST-920

ST-920 rolling submission of BLA to FDA begins

Press Release

(Dec. 18, 2025) Sangamo Therapeutics has initiated a rolling submission of a Biological License Application (BLA) to the FDA seeking accelerated approval of isaralgagene civaparvovec, or ST-920, a wholly owned investigational gene therapy for adults with Fabry disease.

Rolling submission allows for completed modules of the BLA to be submitted and reviewed by the FDA on an ongoing basis rather than waiting for the entire BLA to be submitted at once.

ST-920 has been granted Orphan Drug, Fast Track and RMAT designations from the FDA, Orphan Medicinal Product designation and PRIME eligibility from the European Medicines Agency and Innovative Licensing and Access Pathway from U.K. Medicines and Healthcare products Regulatory Agency. Sangamo expects to complete submission of the BLA to the FDA under the accelerated approval pathway in the second quarter of 2026.

Read full story: bit.ly/st-920

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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Amicus Therapeutics

Galafold stabilizes heart in Fabry women

By Lila Levinson, PhD
FabryDiseaseNews.com

(Oct. 9, 2025) Long-term use of the approved oral therapy Galafold (migalastat) can help stabilize heart and kidney function in women and girls with Fabry disease across different disease severity levels, a new study from U.S. and European researchers reports.

Nearly three-quarters of the female participants in the two clinical trials and their extension studies used for the researchers' analysis had multiple organ systems affected by Fabry. Many also had heart abnormalities, kidney dysfunction, or both. However, Galafold's benefits held regardless of these factors, the data showed.

The researchers say their findings "[challenge] the perception that the burden of Fabry disease is low in females and demonstrates the benefits of [Galafold] treatment in this population."

According to the team, "these data show that females with Fabry disease experience considerable disease severity ... and support the long-term efficacy of [Galafold] for the treatment of" women and girls with the inherited condition.

The study, "Long-term efficacy of migalastat in females with Fabry disease," was published in the Journal of Medical Genetics. While Galafold is approved only for adults with Fabry, these studies also enrolled girls ages 16 and older.

Galafold developer Amicus Therapeutics financially supported the research and collected and analyzed data. Two of the study's eight authors are employed by the biotechnology company.

Read full story:
bit.ly/GalafoldWomen

BioMarin to acquire Amicus Therapeutics

Press Release

(Dec. 19, 2025) BioMarin Pharmaceutical Inc. (Nasdaq: BMRN) and Amicus Therapeutics (Nasdaq: FOLD) announced today that BioMarin has entered into a definitive agreement to acquire Amicus for $14.50 per share in an all-cash transaction for a total equity value of approximately $4.8 billion.

The agreement has been unanimously approved by the Boards of Directors of both companies and Amicus' Board of Directors unanimously recommended that Amicus' stockholders vote to adopt the agreement. The transaction is expected to close in the second quarter of 2026, subject to regulatory clearances, approval by the stockholders of Amicus and other customary closing conditions.

"Amicus, like BioMarin, is a company that has been profoundly dedicated to transforming care for patients with rare diseases since its founding, developing and bringing to market important therapies for individuals living with Fabry disease and Pompe disease. BioMarin's scale of operations, including our global commercial footprint and industry-leading, in-house manufacturing capabilities make the combination of these companies an exceptional strategic fit," said Alexander Hardy, president and chief executive officer of BioMarin.

Read full story:
bit.ly/BioMarin-Amicus

European Medicines Agency committee gives positive opinion on Elfabrio monthly treatment

Move comes after previous rejection of 4-weeks regimen

Press Release

(Jan. 30, 2026) Chiesi Global Rare Diseases, a business unit of the Chiesi Group established to deliver innovative therapies and solutions for people living with rare diseases, and Protalix BioTherapeutics, Inc. (NYSE American: PLX), a biopharmaceutical company focused on the discovery, development, production and commercialization of innovative therapeutics for rare diseases with significant unmet needs, today announced an update on Elfabrio® (pegunigalsidase alfa).

The Committee for Medicinal Products for Human Use (CHMP) of the European Medicines Agency (EMA) has issued a positive opinion recommending approval of the 2mg/kg every-4-weeks (E4W) dosing regimen for Elfabrio in Fabry disease adult patients stable with an ERT (Enzyme Replacement Therapy) treatment. This positive opinion follows the CHMP's re-examination of the company's application for the additional dosing regimen.

"It is our privilege to provide the Fabry community with a safe and effective option, and we are thrilled that the CHMP positive opinion supporting an every-four-week dosing regimen brings us one step forward to further reducing treatment burden in this condition," said Giacomo Chiesi, Executive Vice President, Chiesi Global Rare Diseases.

"We're focused on evolving treatments based on real-world needs, so that people have not just the right care, but care that fits naturally into their lives. By extending the time between infusions, our aim is that people living with this condition can focus on what truly matters, living their lives."

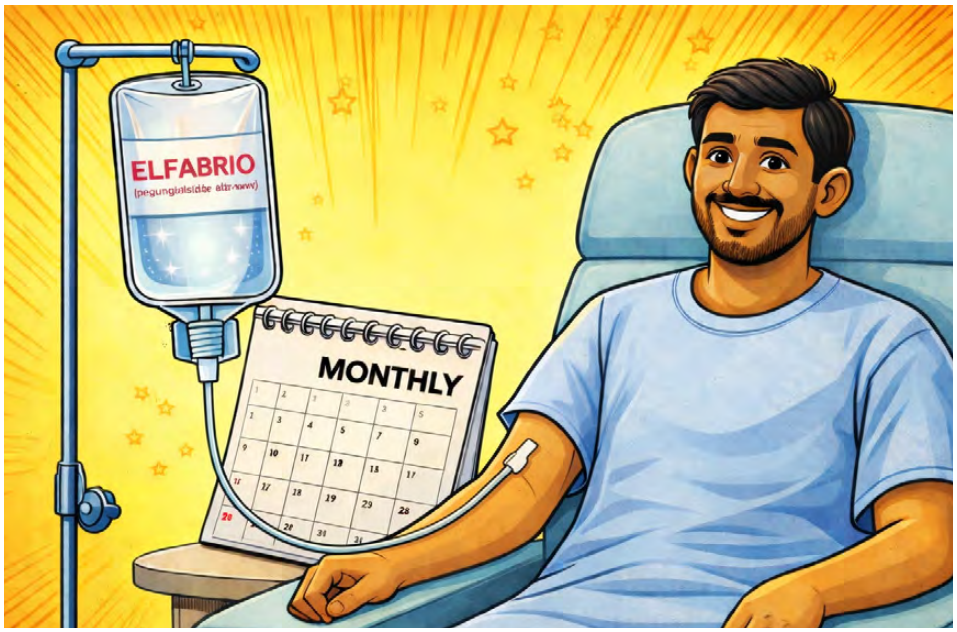
"Expanding the range of treatment options is critical to better meet the needs of people with Fabry disease," said Prof. Aleš Linhart, DrSc, FESC. "The CHMP positive opinion on the every-four-week regimen recognizes the importance of reducing treatment burden for people living with Fabry and their families," said Mary Pavlou, President, Fabry International Network (FIN). "Extending infusion intervals allows therapy to better fit into everyday life, supporting work, study, and family commitments."

The CHMP opinion is based on results from an open-label, switch-over study, BRIGHT (formally PB-102-F50), designed to assess the safety, efficacy, and pharmacokinetics (PK) of the new dosing regimen of pegunigalsidase alfa 2 mg/kg E4W for 52 weeks, and its ongoing open-label extension study CLI-06657AA1-03 (formerly PB-102-F51, with a median exposure of almost 6 years).

Further support is provided by an updated Population Pharmacokinetics model and exposure–response analysis, which leverage data from multiple clinical studies. Protalix will be eligible to receive a regulatory milestone payment of $25 million from Chiesi if the E4W dosing regimen is approved by the EC.

Monthly dosing for Elfabrio is currently not approved in the United States.

Read full story: bit.ly/4Elfabrio



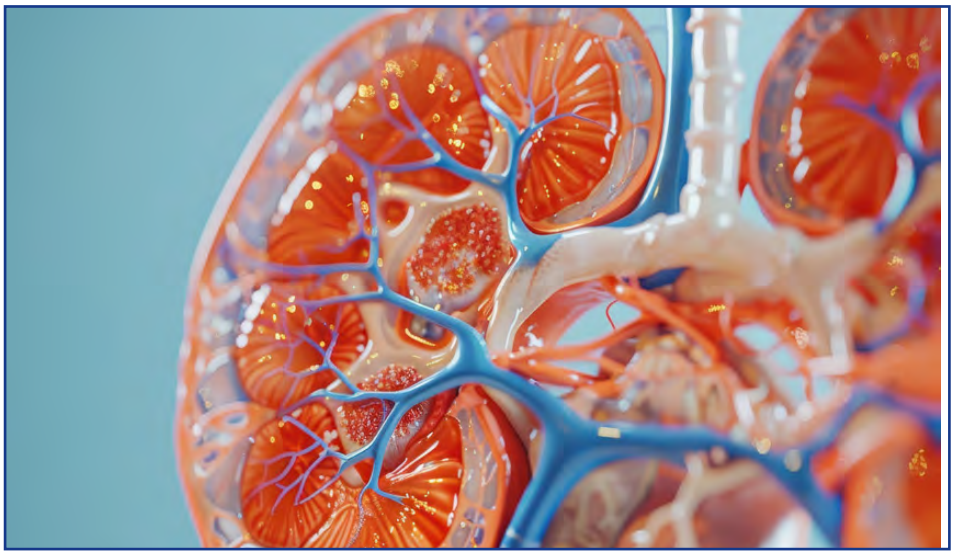
Health Canada approves Elfabrio

(Dec. 18, 2025) Health Canada has approved the enzyme replacement therapy Elfabrio (pegunigalsidase alfa) to treat adults with Fabry disease.

The clearance follows clinical trials showing the treatment was safe and effective. Julia Alton, executive director of the Canadian Fabry Association, called the approval "a powerful moment for the Fabry disease community."

Read full story: bit.ly/FabryCanada

Fabry ERT can limit damage to blood vessels in kidneys



By Marisa Wexler, MS
FabryDiseaseNews.com

(Sept. 25, 2025) Enzyme replacement therapy (ERT) may help limit damage to blood vessels in the kidneys of people with Fabry disease, a new study reports.

Specifically, the study found that people with Fabry who had been on ERT for longer had higher levels of VEGF-165b. This molecule is known to help repair specialized cells in the kidney that are essential for blood vessel integrity.

The study, "Urinary VEGF-A165b mRNA expression in Fabry Disease. Pilot study," was published in *Nefrología*.

VEGF-A165b is a molecule that can help protect small blood vessels. In studies of diabetic kidney disease, it showed a protective effect by helping restore podocytes, which are specialized cells in the kidney that wrap around tiny blood vessels inside filtering units called glomeruli.

Since Fabry patients often have damage in kidney blood vessels, scientists in Argentina hypothesized that VEGF-165b activity in the kidneys may be elevated in this population. To test this idea, the researchers analyzed urine samples from 24 people with Fabry disease and 24 healthy volunteers.

The scientists measured VEGF-165b messenger RNA (mRNA) levels in the urine samples. mRNA is an intermediary molecule made when genes are read to make protein. Since VEGF-165b mRNA is used to make VEGF-165b protein, higher mRNA levels indicate higher protein levels.

The scientists found that VEGF-165b mRNA levels tended to be higher in urine from Fabry patients, but the difference was not statistically significant (meaning it's mathematically plausible that this difference is random chance). The researchers stressed that this was a relatively small study, noting that larger studies may be needed to find statistically meaningful results.

Nine of the Fabry patients were taking the ERT Fabrazyme (agalsidase beta). Statistical analyses of data from these patients revealed a significant correlation between time on ERT and levels of VEGF-165b mRNA. In other words, patients taking ERT longer tended to have higher VEGF-165b mRNA levels.

Since VEGF-165b is associated with blood vessel repair, the researchers speculated that this finding might reflect kidney vessel repair in Fabry patients who've been on long-term treatment. "Probably in patients with longer treatment time there is a decrease in [Fabry-related] damage," the researchers wrote.

Read full story: bit.ly/ERTkidney

ERT may protect heart function after kidney failure

By Steve Bryson, PhD
FabryDiseaseNews.com

(Oct. 30, 2025) Enzyme replacement therapy (ERT) preserved or improved heart function in two men with Fabry disease who had already experienced kidney failure and were undergoing renal replacement (dialysis or kidney transplant), a new case study reports.

The study provides evidence that ERT, the standard treatment for Fabry disease, can still provide benefits to the heart even when the kidneys are severely impaired.

"Patients with Fabry disease who are on renal replacement therapy may benefit from enzyme replacement therapy," researchers wrote.

Read full story: bit.ly/ERTheart

MORE STORIES

Can CHO bioreactors be used for plant-based protein production?

A company developing protein therapies otherwise challenging to make by traditional means is pursuing moss production to produce the recombinant version of human glycoproteins, with its lead candidates targeting Fabry disease (RPV-001) and glomerulopathies (CPV-104).

bit.ly/Bioreactors

Fabry linked to small fiber neuropathy in twins

New research shows that Fabry disease can affect the nerves of females—even in childhood—earlier and more clearly than once thought.

bit.ly/FabryFiber

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