SUMMER/FALL 2025

GALACTOSEMIA GAZETTE

Galactosemia Foundation

Linked for Life

President's Message

BY: NICOLE CASALE

As we head into the last few months of 2025, I'm reminded of how powerful our community is—whether we're together in person or connecting virtually across the miles. Each event we host, from a local museum outing to an online workshop, strengthens the bonds that make this community so unique.

This fall and winter, we're thrilled to offer a mix of outreach opportunities. Our goal has always been to meet families where they are—sometimes that means in person, and other times it means a Zoom room filled with familiar, supportive faces. Both matter. Both help us grow.

As we plan ahead for 2026 and our next conference, I encourage you to stay connected, join an event, and lend your voice. Every conversation, every shared idea, every volunteer effort makes a difference. Together, we continue to move forward with resilience, purpose, and hope.

Let's keep building these connections—near and far.

MARK YOUR CALENDAR: July 16 - 18, 2026
Galactosemia Conference in Aurora, CO

Join the Citizens Health Registry - Your Voice Matters

The Galactosemia Foundation is excited to announce the launch of our new Citizen Registry, a powerful tool to unite patient and family voices, gather vital data, and drive change at every level — from research to policy.

By signing up, you'll help:

- Advance research by giving scientists access to real-world patient experiences.
- Strengthen our advocacy efforts with the FDA, lawmakers, and industry partners.
- Build a clearer picture of the burden of disease, helping accelerate treatments and improve care.
- Replaces placebo arms of studies.

Every entry matters — whether you are a patient, caregiver, or family member. Your participation can directly influence clinical trial design, biomarker recognition, and access to therapies. This data can replace placebo studies in children and adults.

Sign up today and be part of the change! Join the Citizen Registry here: www.citizen.health/partners/galactosemia-foundation

📅 Save the Date: We'll be hosting a webinar to share updates, answer questions and show how registry data will be used to help our community. Details will be shared soon — but signing up for the registry now ensures you'll be the first to know.

Together, we can make sure the Galactosemia community is heard loud and clear.

Community Spotlight

We Want to Hear From You!

We're launching a Community Spotlight series and would love to feature you! Whether you're a parent, sibling, individual living with galactosemia, grandparent, or caregiver—your journey matters. Sharing your story can help:

✓ Raise awareness
✓ Support newly diagnosed families
✓ Highlight the strength and resilience of our community

If you're open to being featured in our newsletter, website, or social media, please fill out our short interest form below. Stories can be written, audio, or even video—whatever you're comfortable with!

Share your story here: https://forms.office.com/r/VbhNM952CX

Giving Tuesday

This Giving Tuesday, we're coming together as a community to make a difference for everyone affected by galactosemia. While each of our journeys look different, we share the same hope—for more awareness, more research and more support for our families.

Your participation is vital. Every dollar raised and every story shared helps shine a light on galactosemia, strengthening our collective voice and fueling progress. Together, we can ensure that our community feels seen, supported and empowered.

Let's show the power of what we can achieve when we all come together. Please join us this Giving Tuesday - December 2 - to raise funds and awareness for the good of our galactosemia community. More info to come soon through email and on our social media.

Join Us for a Favorite Things Gift Exchange

What It Is: Each participant picks a favorite item (or small bundle of items) they love—valued at or under $20—and mails it to another participant. At the Zoom meetup, everyone opens their gifts and shares why they picked their "favorite thing." Gift must be mailed by December 2. Sign up here.

Connect with Us

Follow Galactosemia Foundation on Social Media and visit our website.

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Funded Research Updates

The Galactosemia Foundation provides funding for scientific and medical research related to the identification, treatment, and therapies related to classical galactosemia. The goal of this research initiative is to improve the lives of people with galactosemia. Here are updates from funded research projects currently underway.

Granulosa Cell Gene Therapy for Classic Galactosemia - Dr. Kent Lai
While the underlying disease mechanisms of Classic Galactosemia are not fully understood, none will argue that the disorder is caused by deleterious mutations in the GALT genes, which result in the near-/complete absence of cellular GALT enzyme activity. Therefore, therapeutic approaches that offer to restore the missing GALT activity in patient cells represent a rational and promising therapeutic strategy. Dr Elias (Co-PI) at Cleveland Clinic and I have been exploring a non-viral mediated gene therapy that employs nanoparticles to specifically target the granulosa cells in the ovaries and delivers a functional GALT gene. This targeted approach is advantageous because for female patients who do not have complications other than ovarian insufficiency, they would not need any therapeutic agent that will otherwise be deployed to their brain or other organs. In this Research Grant award, we would conduct proof-of-concept studies to animal models of Classic Galactosemia so that we can fully evaluate the efficacy of this experimental therapy in a living mammal, which is required to attract the biopharma industry to bring this therapy to clinical stage.

Functional Outcomes from Pediatrics to Adulthood in Classic Galactosemia - Dr. Dave Jensen and Dr. Nancy Potter
This study is a follow up to an original study conducted in 2005 and 2007, in which Dr. Potter tested 33 children with classic galactosemia (CG), ages 4-17, in their homes.

Now, 18-20 years later, we want to understand how the same individuals are doing as adults in regard to speech, language, cognition, motor skills, employment, and life skills.

The original study was foundational for learning about speech, language, cognition, and motor development in CG and developing the universal criteria for diagnosing childhood apraxia of speech.

The current study will provide important detail about the longitudinal picture of CG and examine the relationship between childhood and adult strengths, challenges, social engagement, employment, and living independence. These results will be important for answering questions about whether speech, language, cognitive, and motor skills improve or decline over time, for advocating for educational therapy support during childhood and adolescence, and providing baseline information for long-term outcomes for biologic and therapy interventions.

If you were in the original study, please let us know your current contact information and please say “Yes, I’d love to participate” when we contact you.

Funded Research Updates (continued)

Phenotyping of movement disorders in classic galactosemia, moving towards guidelines for individualized treatment - Dr. Tom de Koning and Dr. Estela Rubio Gozalbo

We are very happy that we have been awarded a grant from the Galactosemia Foundation to start our research project on involuntary movements in galactosemia. Many patients suffer from symptoms such as trembling, shaking, jerky movements or have problems with balance and coordination, but these symptoms are often poorly recognized and classified by most doctors. Using video registrations and neurophysiology measurements, we will study in detail these involuntary movements and combine this with how the involuntary movements impact daily life of galactosemia patients. Together this information will be used to make a treatment guideline for movement disorders in galactosemia that will aid physicians to recognize and treat the involuntary movements.

Navigating Primary Ovarian Insufficiency (POI) in Galactosemia - Dr. Judy Fridovich-Keil

Primary ovarian insufficiency (POI) is the most common long-term complication experienced by girls and women with classic galactosemia, but it remains poorly understood. With new support from the Galactosemia Foundation, we are extending our ongoing longitudinal study of galactosemia to focus more attention on 2 important aspects of outcome and intervention related to ovarian function: (1) puberty and hormone replacement therapy (HRT), and (2) pregnancy. These are extremely important topics because we have learned from our ongoing study that not all young women with galactosemia and POI are receiving adequate doses of HRT to protect their long-term bone and heart health. We have also learned that many young women with galactosemia and POI who would like to have a baby have been advised they are infertile and so do not even attempt pregnancy – yet those who do are sometimes successful.

Specifically, we plan to distribute a new survey to girls and women enrolled in our ongoing study that asks, in detail, about experiences with HRT and/or pregnancy. If you are already enrolled in our study, please watch your email for the new survey invitation.

If you are not yet enrolled in our study but might want to get involved, please contact Judy Fridovich-Keil at jfridov@emory.edu or 404-727-3924. Thank you! The more people participate, the more meaningful the study will be. We can't wait to get started!

Mark Your Calendar: Upcoming Events

Visit Galactosemia.org for more information and to stay up-to-date on all events.

2025

- **November 13**: (Virtual): Adult Financial Literacy Workshop- Part 1: Savings & Spending Plans
- **December 6**: Dallas, TX; Perot Museum of Nature & Science
- **December 14**: (Virtual): "Favorite Things" Gift Exchange (Optional)

2026

- **January 16**: Los Angeles, CA; The Aquarium of the Pacific
- **March 28**: Troy, NY; Silent Disco Fundraiser-Glow for Galactosemia
- **April 26**: Hamden, CT; Denise D'Ascenzo Foundation's Walk to Fight Rare Diseases

Recently Published Research: A Pilot Study of Bone Marrow Transplantation in a GALT- Null Rat Model of Classic Galactosemia
By: Dr. Judy and team

Our team recently published a paper entitled "A Pilot Study of Bone Marrow Transplantation in a GALT- Null Rat Model of Classic Galactosemia" in the journal JIMD Reports. Here is a summary of what we learned:

- Successful bone marrow transplant (BMT) with GALT-positive donor cells, but not GALT-null donor cells, restored strong GALT activity to the red blood cells of the GALT-null recipient rats.
- Successful bone marrow transplant (BMT) with GALT-positive donor cells, but not GALT-null donor cells, also normalized red blood cell gal-1P in the GALT-null recipient rats.
- However, successful BMT with GALT-positive donor cells did not restore detectable GALT activity to liver or brain, and also did not correct other metabolites in blood, or any metabolites in liver or brain.
- We therefore concluded that, unless some altered procedure could allow bone-marrow-derived cells improved access to solid tissues like liver and brain, BMT may not be a good option for intervention in galactosemia.
- Of course, this study, like all studies, had strengths and limitations, and these are explained in detail in the Discussion section of the paper.

Get Involved with the Galactosemia Foundation

The Galactosemia Foundation is completely volunteer-led. There is always room for more and would love to have members of our community play a bigger role in the Foundation. Check out the opportunities below and let us know if one sounds like a good fit for you!

How You Can Help

We have a few exciting volunteer opportunities where your skills and experience can make a real difference:

1. Mock Employment Interviews
   - Help community members build resumes and practice interview skills.
   - We're looking for volunteers with hiring experience who can perform mock interviews and provide constructive feedback.
2. NEON Support
   - Assist with our CRM (customer relationship management) and fundraising system to help us streamline outreach and donor management.
   - Anyone familiar with NEON's tools can help us maximize their impact.
3. Mentoring Program
   - Share your experience and guidance with individuals in our community navigating school, work or life transitions.
   - Provide encouragement, advice, and a friendly ear for those looking for support.

Your time and expertise can help our community grow stronger—thank you for considering lending a hand!

Interested in being part of the board or a committee click here.

Interested in being a virtual volunteer click here.

A Message from Dr. Judy and Team: Study Participants Needed

Researchers at Emory University in Atlanta, Georgia, are conducting an observational, longitudinal study of long-term outcomes in classic galactosemia (CG).

They are looking for people with classic (or clinical variant) galactosemia (CG) to participate. The more people participate, the more powerful the study will be.

With support from the Galactosemia Foundation, we are excited to expand the focus of our study on ovarian function in CG, including a new survey about ovarian function.

This study has been approved by the Emory University Institutional Review Board (IRB Protocol 00024933; Principal Investigator: Judith Fridovich-Keil, PhD).

About the Study

Participation is easy and non-invasive -- no extra needle stick! There is no study-related costs to you or your insurance company and no travel required.

Results from this study may answer important questions about the natural history of galactosemia, now expanded to include what, when, and how much hormone replacement therapy (HRT) is best for girls and women with CG, and how women with CG who wish to become mothers have navigated the journey.

Who is eligible?

Anyone conversant in English with a confirmed diagnosis of classic galactosemia may be eligible to participate as a "case."

Siblings who do not have galactosemia may be eligible to participate as "controls."

What's involved?

- Informed consent process: a Zoom call with the study PI (Judy) and completing an online form
- A set of online surveys to complete covering a range of outcome domains
- A request for authorization to obtain relevant medical records
- We may ask for a saliva sample to allow for genetic studies. If you live in North America, when you have a clinical blood draw, we may also ask if the phlebotomist would please collect a small extra tube of blood for research. There is no cost to you or your insurance company for this procedure, and no extra needle stick. We will not ask for blood from controls, or from cases more than once a year
- Girls who donate a blood sample may receive free analyses of hormones related to ovarian function

Interested in joining the study? Please email Judith Fridovich-Keil at jfridov@emory.edu with questions or comments.