SUMMER 2023

GALACTOSEMIA GAZETTE

Galactosemia Foundation

Linked for Life

President's Message

BY: NICOLE CASALE

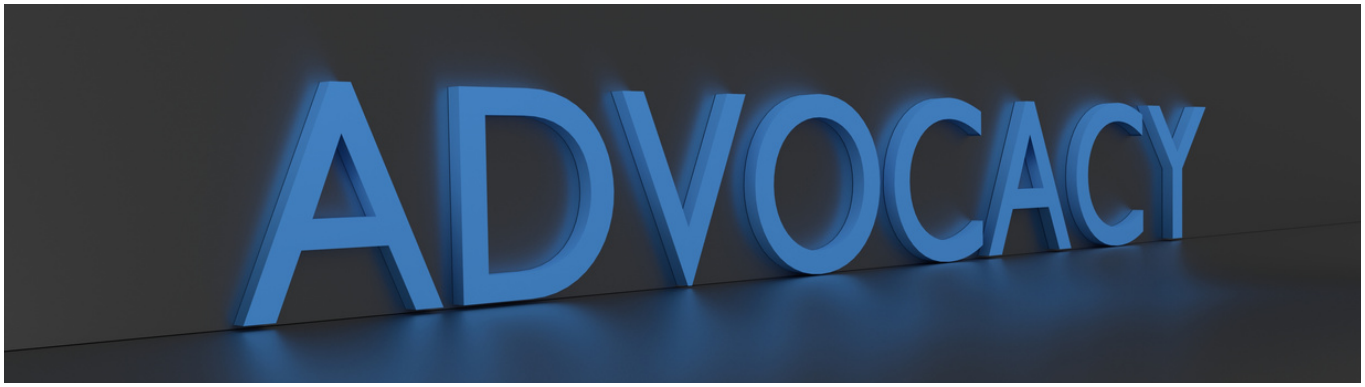
To my fellow galactosemia advocates,

I am in awe of our community's efforts over the last few months as we have shared our stories and raised our voices to help the FDA understand the want and need for the first-ever treatment for galactosemia. We have worked together to highlight the tremendous burden of this disease, the improvements we've seen in children who were taking govorestat in the clinical study, and the heartbreak for children on placebo or who weren't able to be in the study at all. Sharing our personal struggles so publicly can take an emotional toll; it can be hard to relive some of our children's most difficult experiences and put them into words that others can understand. Each one of you is strong and resilient, and I am so grateful for your work on behalf of this community.

Since learning of the govorestat clinical trial data, we have:

- Created a Change.org petition that has garnered more than 10,000 signatures
- Shared our stories on social media, with reporters, and via op-eds and podcasts
- Written letters to the FDA
- Met with policymakers
- Published and promoted the historic "Voice of the Patient" report, summarizing our perspectives from the externally-led patient-focused drug development meeting

We know advocacy can be time-consuming and overwhelming, but we are doing everything in our power to make a difference. It remains more important than ever for our community to continue to rally behind this important cause. Check out the Advocacy article in the newsletter to learn more about our efforts and how you can get involved.



WE NEED YOU: ADVOCATING FOR A TREATMENT

The Galactosemia Foundation is urging U.S. Food and Drug Administration (FDA) to incorporate the experiences and perspectives of people with galactosemia in the agency's review of govorestat (AT-007), the potential first-ever treatment for our rare genetic disease.

While the ACTION-Galactosemia Kids trial data did not quite meet statistical significance, Applied Therapeutics believe it has demonstrated compelling evidence of clinical benefit and a favorable safety profile in patients with Galactosemia. They were recently granted a meeting with the FDA this summer to discuss a potential New Drug Application (NDA) submission.

In the meantime, we are working tirelessly to tell our story, in hopes that the FDA considers patient and caregiver perspectives when evaluating risk-benefit for potential new treatment options.

We are asking the community to help advocate for a treatment by sharing their personal stories related to galactosemia and by helping educate friends and family and asking them to advocate for our community. We have an Advocating for Treatment page on our website where you can find information and action items. We will also share a few in the newsletter, so read on to see how you can help.

Why are we advocating?

If you are new to our galactosemia community or a friend or family member is asking you to advocate, you may be asking "for what" - so here is the background and resources to get you caught up.

The Voice of the Patient Report

The Voice of the Patient report, published jointly with NORD, provides a comprehensive overview of patient and caregiver experiences with this rare metabolic disease.

Read and share
Voice of Patient Report

Why are we advocating?

The Hill Op Ed: The FDA is ignoring patients in clinical trials
By: Brittany Cudzilo, Galactosemia Foundation VP

"Imagine having two daughters who both have a rare disease and watching them struggle — to be understood, to remember things, to learn, to be comfortable in social settings, to follow one-step directions, to be able to bond with friends and family.

Now, imagine a clinical trial showed promise for your older daughter. You notice a remarkable difference in her daily living, and suddenly, your home is more peaceful, and she is starting to thrive. The treatment is potentially life-changing. You now have hope for your older daughter and her younger sister.

But then, the medicine is potentially at risk of not even being reviewed by the U.S. Food and Drug Administration (FDA), let alone approved. As a parent, what would you do? What would you say to the FDA?"

Read and share
The Hill Op Ed

Change.Org petition: PaveTheWayFDA - Galactosemia Community Urges FDA to Incorporate Voice of the Patient in Review of Potential First Treatment for Patients with Life-Altering Rare Disease

"Based on the tremendous burden of disease and the compelling evidence that govorestat (AT-007) may offer galactosemia patients a better chance of an independent life, the galactosemia community urges the FDA to review and approve this medicine as quickly as possible. We know the FDA is dedicated to listening to patients and advancing safe and effective new medicines that will benefit patients, and we hope to partner with the FDA to advance this mission on behalf of the galactosemia community."

Our Petition currently has over 10,000 signatures, which is impressive for a rare disease community. Please help us get the number to 15,000 by signing and sharing the petition with your friends, family and social networks.

Sign and share
Change.org petition

Why are we advocating?

SHARE YOUR STORY
Everyone in our community has a story to tell.

Some can talk about their experiences with the clinical trial. That may include the benefits experienced since their child has been on drug, or the declines that continue to happen if their child was on placebo.

Some can talk about what a treatment would mean to their family. How would life improve if you saw improvements in cognition and other daily living skills and reduction of tremor?

If you're not directly impacted by galactosemia, share the stories of your family, friends and neighbors. Are you a physician? Speak up for your patients. A teacher? Tell the story of your student. A rare disease supporter? Share the importance of treatment. Your voice matters.

Regardless of your connection to galactosemia, you can help us implore the FDA to take our stories, struggles and lives into consideration as they make decisions that directly impact our community. Ready to get started but not sure where to tell your story? Check out these links to get started:

Patients Rising
Share your story with a group of passionate passion advocates who believe the patient voice should be central to healthcare and barriers to access should be removed.
https://patientsrisingstories.org/get-involved/your-patient-story/

Congress
Contact your members of Congress to share your story,

- Contact the Rare Disease Caucus

The Galactosemia Foundation
Email us at outreach@galactosemia.org with quotes and photos or a short video (think reels) to share your story about the burden of galactosemia and the need for a treatment (we will then share on our social media channels).

Thank You Beatrice Ortego!

As Beatrice Ortego retires from the Research committee, we want to recognize and thank her for her generous time. She has been an integral player through the years, providing her scientific expertise during the review process and ultimately guiding the Foundation in the research process. Thank you, Beatrice!

MARK YOUR CALENDAR: July 18 - 20, 2024
2024 Galactosemia Conference in Charlotte, NC

Looking for adults with classic or clinical variant galactosemia - 30 years and older

WHAT IS THE GOAL OF THE STUDY?

Most of what we currently understand about classic and clinical variant galactosemia (CG/CVG) comes from studies involving children and young adults. The goal of this study is to learn from the experiences of maturing adults with CG/CVG. Do old challenges get better or worse? Do new challenges appear? Our goal is to gather information from as many adults as possible, age 30 and over, living with CG/CVG to answer these questions.

WHO IS ELIGIBLE TO PARTICIPATE?

Any adult, age 30 or older, with a confirmed diagnosis of CG/CVG and the ability to complete an online or hard copy consent, and online or telephone survey, may be eligible to participate. An adult who is not their own legal guardian may still participate with the assistance of their legal guardian.

WHAT WOULD WE ASK YOU TO DO?

Volunteers will be asked to read a consent form, or have it read to them, and ask questions if they are unsure of anything. After completing the consent form, each participant will be sent an email with a link to an online survey that should take about 10 minutes to complete. That's it! Participants who prefer can have the survey administered over the telephone.

WHO IS CONDUCTING THE STUDY?

This study is funded by the Galactosemia Foundation and is conducted jointly by three teams of researchers, two in the US and one in the Netherlands. One team is from Boston Children's Hospital and Harvard Medical School in Massachusetts (lead by Gerard Berry, MD), one is from Emory University School of Medicine in Georgia (lead by Judith Fridovich-Keil, PhD), and one is from Maastricht University Medical Center in the Netherlands (lead by Estela Rubio-Gozalbo, MD, PhD).

QUESTIONS? MIGHT WANT TO JOIN?

Please reach out to the person listed below who is located closest to you:

- Judy Fridovich-Keil in Atlanta, GA, USA (jfridov@emory.edu)
- Debbie Fu (for Gerry Berry) in Boston, MA, USA (yuting.fu@childrens.harvard.edu)
- Estela Rubio-Gozalbo in Maastricht, the Netherlands (estela.rubio@mumc.nl)

Looking Back: GLOW 2023

A sincere thank you to everyone who participated in GLOW for Galactosemia this year. It was a huge success, raising more than $50,000, $12,000 of which will go toward the Paul P scholarship for conference next year. We had over 1,000 participants, including more than 40 galactosemia families. We are excited to see what the future holds for GLOW and our community.

A Notice to Women with Classic Galactosemia Who Take Birth Control Pills for Hormone Replacement Therapy

On July 13, the US Food and Drug Administration (FDA) approved Opill (norgestrel) as a non-prescriptive, over-the-counter birth control pill. For women with typically functioning ovaries who seek to avoid pregnancy, Opill may offer a convenient alternative to prescriptive birth control pills. However, for women with classic galactosemia who take prescriptive birth control pills that contain estrogen to achieve effective hormone replacement, Opill is not a good alternative, because Opill contains progestin and does not contain estrogen. **If you have questions about your hormone replacement therapy options, please consult your personal healthcare provider.**

Judith Fridovich-Keil, PhD
Jessica Spencer, MD

Connect with Us

Follow Galactosemia Foundation on Social Media and visit our website.

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Scott Saylor, Treasurer/Fundraising
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