WINTER 2023

GALACTOSEMIA GAZETTE

Galactosemia Foundation

Linked for Life

President's Message

BY: NICOLE CASALE

Hello Galactosemia Community! I can't believe it's been about ½ a year since we were all together, and while 2024 conference seems so far away, the Foundation is now gearing back up to start planning. So mark your calendars for July 18-20, 2024 and we'll see you at the Embassy Suites in Concord, North Carolina!

Reflecting on conference, I would say it was another success! It was a smaller turnout than we have had in the past, we were sad to have missed so many familiar faces. COVID definitely put a damper on so many things, on top of that there were some kinks thrown in as we attempted our first Hybrid Event. But, for a moment we were able to forget all that and enjoy and learn from each other and the experts. We ate all the safe ice cream, played some fun games, danced the night away, and our talented community did not disappoint at "Talent Together."

A special thank you to Natalie Whittington and her family for putting on an amazing talent show. To Tracey Miller and Nick Casale for running the "No Whey Café." To the Elliot and Shepard families for running another successful age group program, and to the Board and everyone else who made it such an incredible event.

We don't know what the future will bring, but we do know how remarkable our community is, and while we are rare we are always Stronger Together!

Sponsor Highlight - Applied Therapeutics

Applied Therapeutics Sponsors the Galactosemia Foundation Website Redesign, Education Materials and GLOW for Galactosemia Family Fun Race Event

Thank you to Applied Therapeutics for sponsoring the Galactosemia Foundation's updated website (www.galactosemia.org) and the Newly Diagnosed Toolkit for Families (https://www.galactosemia.org/resources/toolkit/) Both feature actual photos of people living with Galactosemia!

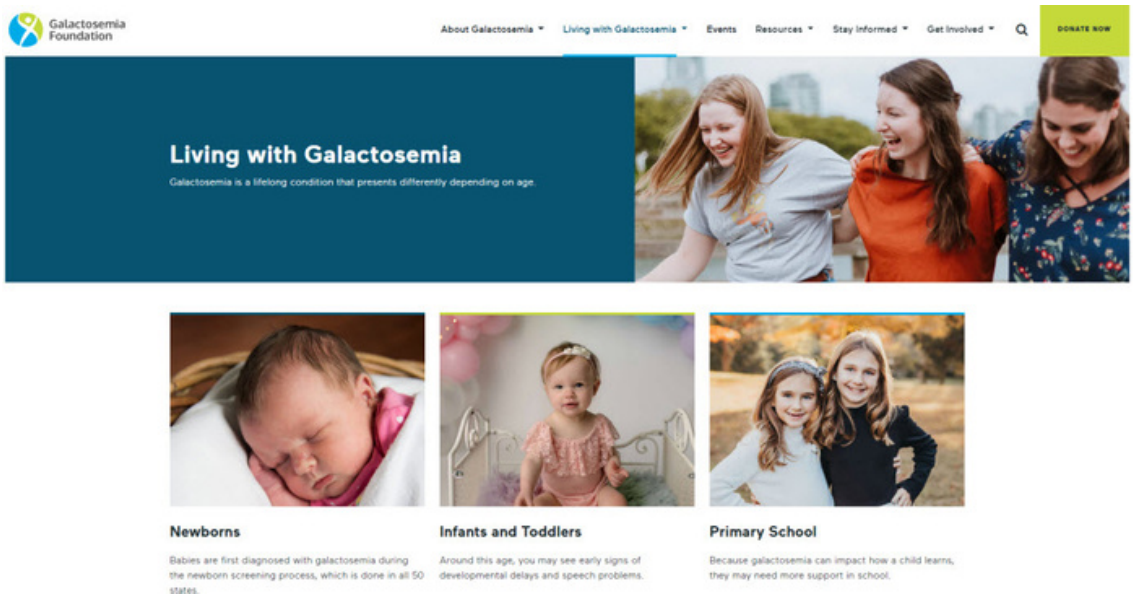
Education and awareness of Galactosemia is essential for parents, healthcare providers and those living with Galactosemia. These new resources will help families better navigate their loved one's Galactosemia journey, connect with the community, empower them to actively engage with their medical team, and be an advocate for the community.

Did you know that Galactosemia Together, an educational website for families, patients and healthcare professionals, is celebrating two years since it's launch?! This educational resource was co-created with Applied Therapeutics and the Galactosemia community, and it continues to be a go-to resource for families, those living with Galactosemia and healthcare providers. Check out the site at www.galactosemia.com

Come and meet some of the Applied Therapeutics team at the GLOW for Galactosemia Family Fun Race Event, taking place Sunday, March 5, 2023 in Knoxville, TN. Click this link to register and for more information: https://runsignup.com/Race/Events/TN/Knoxville/GLOWforGalactosemia2019

If you can't make the event and/or are participating virtually, you can connect via email with Dottie Caplan, SVP Patient Advocacy and Engagement – Applied Therapeutics at dcaplan@appliedtherapeutics.com.

A New Look for the Foundation



If you haven't been to the Galactosemia Foundation website in a while, now is the time to go take a look! Thanks to a sponsorship from Applied Therapeutics we were able to update existing and create new assets. We worked with Cullari Group on several initiatives including a refreshed logo, a newly diagnosed toolkit and a redesigned website.

The website still has many of the things our community depends on, like the current dietary guidelines. But it also has more information based on different ages to help our community through the various challenges associated with each stage of life.

One thing you'll notice on our website and other resources is actual pictures of our community members. Thank you to everyone who answered the call on social media and the newsletter to send us hi resolution photos. It definitely makes a difference in the authenticity of our materials. If you didn't get a chance but would like your child (or yourself – if you have galactosemia) to be featured in upcoming social posts, newsletters or website content please email hi resolution photos to media@galactosemia.org.

The Toolkit for newly diagnosed families is available on the Resources page and is meant to be a high level introduction to families experiencing the diagnosis for the first time. Check out the new website: Galactosemia.org. You can also download the online toolkit here.

Galactosemia Foundation's Advocacy Committee Helps Pass HEART Act

In January of 2022, we joined the Haystack Project addressing Congress to implement important changes in the drug review process at the FDA for rare disease.

The HEART (Helping Experts Accelerate Rare Treatments) Act will ensure simple commonsense changes to include experts that understand rare disease. This has been a pivotal time as Applied Therapeutics' investigational drug for galactosemia, AT 007, is currently under the review process at the FDA. The HEART Act addresses many issues that the drug trial faced in the last year. This act will benefit future review of treatment options for galactosemia and other rare diseases.

On Dec. 29, 2022, with the passing of the Omnibus, the HEART Act was passed, which will amend the Federal Food, Drug and Cosmetic Act to improve treatment of rare disease and conditions. Details include:

- Requires a study on sufficiency and use of FDA mechanisms to incorporate patient/clinician perspective in FDA processes related to applications for drugs for rare diseases & conditions
- Calls on the FDA to be required to develop an annual report on progress of rare disease drug applications
- Requires FDA host a public meeting to address approaches to increasing and improving engagement with rare disease or condition patients, groups representing such patients, rare disease or condition experts, and experts on small population studies, in order to improve the understanding with respect to rare diseases or conditions in terms of patient burdens, treatment options and side effects
- Directs a review of the European Union's best practices for approving rare disease drugs
- Includes experts in small population science studies

Thanks to all our galactosemia families and patients who supported our advocacy efforts. We could not do this without you. Also, a special thank you to Senator Casey and Scott and Representatives Tonko and McKinley for representing rare diseases and working hard to pass this act.

Celebrating Rare Disease Month

February is rare disease month and we are raising funds for the Galactosemia Foundation to support rare disease research and education related to Galactosemia! You can help!
Please Text: Rare to 71777
You will be given option to donate or become a fundraiser.
Once you become a fundraiser you can also choose to form or join a team!
You will also be given the option to create your own Team page!
All you have to do is get the word out to TEXT Rare 71777.

RARE DISEASE DAY®

GLOW FOR GALACTOSEMIA: YEAR 6

GLOW for Galactosemia is an event hosted in Knoxville and available across the United States that raises funds for the Galactosemia Foundation. Last year, GLOW was represented in 41 states with over 1,000 people participating, as families and friends of someone with Galactosemia showed support by being part of the virtual option.

This year, 1/4 of the funds raised will go straight into the Paul P scholarship fund to help families come to the 2024 conference, and the other 3/4 will be placed in the general fund to help with outreach efforts and research projects.

GLOW has raised over $150,000 in the past 5 years, in large part because families in our community have shared their Galactosemia story and invited their friends and family to help support our foundation through virtual participation.

Virtual participants receive a package in the mail with a glow in the dark (ink) t-shirt, a sweatshirt (while supplies last), and other swag items! For just $30 you and your friends can help support the Galactosemia Foundation!

The last day for virtual sign ups is Feb. 3. If you come to Knoxville, TN there will be a Galactosemia families dinner Saturday night and the GLOW event on Sunday afternoon. All people with Galactosemia GLOW for FREE.

Email Brittany.cudzilo@galactosemia.org to learn how to register.

Connect with Us

Follow Galactosemia Foundation on Social Media and visit our website.

Linked for Life

Contact The Board

Nicole Casale, President
Brittany Cudzilo, Vice President/Outreach
Scott Saylor, Treasurer/Fundraising
Jodie Solari, Communications
Cari Miller, Secretary
Keith Topper, Board Member at Large
Kelley Foley, Board Member at Large

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)

Research Participants Needed

Researchers at Emory University in Atlanta, Georgia, are conducting an observational, longitudinal study to define the timing, prevalence, modifiers, and potential changes over time of long-term complications in classic galactosemia.

Results from this study may answer important questions about outcomes experienced by patients and may aid the development of new treatments. Please help!

What's Involved?

- After you complete the informed consent process we will ask you to fill out a set of online surveys that ask questions about your or your child's medical and family history. Then, every 6 months through age 5, and annually after that, we will ask you to send updated information using online follow-up surveys. We will also ask for authorization to request medical records from relevant healthcare providers.
- We may ask for a saliva sample to allow genetic studies. When you or your child are scheduled to have a clinical blood draw, we may ask if the doctor could please collect a small extra tube of blood for research. If both you and the doctor agree, we will send you a kit with the tube, instructions, packing materials, and a pre-paid shipping label. There is no cost to you or your insurance company for this procedure, and no extra needle stick.
- Participants who donate a blood sample may receive free GALT genotype analysis, and girls who donate a blood sample may also receive free analyses of hormones related to ovarian function.

Other Information

- Participation is easy and non-invasive - no extra needle stick, really!
- No study-related costs to you or your insurance company.
- No special travel -- all required components can be completed remotely.
- Subjects who complete all their surveys will be compensated $50 for their first year of participation, and $25/year for up to 4 additional years of participation.

Questions or ready to join?

- ·Please email Judith Fridovich-Keil at jfridov@emory.edu, or contact our research team at galactosemia@emory.edu

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

When Vegan is Not Dairy-Free

Up until recently, a food product that was vegan was also dairy-free. After all, vegan has traditionally meant the food does not contain any products derived from animals; and dairy-free has typically meant the food does not contain dairy, including milk, milk proteins and other milk ingredients. Importantly, dairy has always come exclusively from animals. Therefore, a food that did not contain product from animals (i.e., a food that is vegan) could not contain dairy and so it must be dairy-free. Thus, seeing the term vegan on food packaging was an easy way to determine the food was safe for the Galactosemia diet.

Unfortunately, it is no longer that simple. Companies such as Brave Robot have begun to manufacture milk proteins in laboratories without the use of any animals. As such, these companies can label their products as vegan even though they contain dairy. This loophole poses a threat to people with Galactosemia who rely on the term vegan as a shortcut for ingredient label-reading.

The exact process that companies like Brave Robot use to create their products is scientifically complex. In a nutshell, Brave Robot encodes specific bacteria with the DNA sequence of whey protein to create proteins that are “bio-identical” to their naturally occurring counterparts. Ultimately, these companies are able to label their products as vegan because the production processes do not involve animals.

There are several reasons why companies are using these kinds of production processes. The companies claim these processes are better for animals (and for the environment) than traditional milk production processes. Being that these processes - by design - exclude animals, it is hard to argue with that. However, what should not be overlooked is that companies want to be able to label their products as vegan. There is a sort of positive aura that surrounds the enticing, alluring term vegan. Consumers typically associate those foods with being organic, all-natural, free of harmful ingredients, and just generally healthier than their non-vegan counterparts. Companies want to be able to capitalize from the ideas that are elicited by the term vegan.

One possible solution to this loophole is for the FDA to implement a new term for this manufactured dairy. This term would be posted in a food product’s ingredient list, just as all other ingredients should be. Alternatively, the FDA could create a new symbol to use on products that contain these types of ingredients–one that is easily identifiable, like the green “USDA Organic” emblem or the encircled “K” that represents a Kosher designation.

While these kinds of advancements in agriculture can create challenges, they can also be beneficial to consumers. For example, without agricultural advancements, there would be no multibillion-dollar plant-based milk industry that the galactosemia community has come to depend on. As companies continue to test the limits at the intersection of agriculture and technology, similar problems may continue to arise and exemplify the importance of label-reading, regardless of how the product is marketed.