

# GALACTOSEMIA GAZETTE

OFFICIAL NEWSLETTER OF GALACTOSEMIA FOUNDATION



## President's Message

BY: KELLIE WILCOX, PRESIDENT

Well, 2020 is ending and I think we can all say it has been a 'one of a kind' year! While many things have happened in each of our lives during this year, there has also been a lot happening in the Galactosemia world.

Applied Therapeutics has been making a name in the Galactosemia community. They started a drug trial this year with hopes of making life-altering changes in the Galactosemia community. Due to COVID-19, the trial was put on hold. There is more info on page 2 of the newsletter. We are so thrilled to see where this trial goes.

For the first time ever, we had a virtual Galactosemia Conference. One of the main reasons for the conference is social interaction; for parents, those with Galactosemia, colleagues, etc. Instead of cancelling the conference we made adjustments and held our first ever virtual conference. Despite the short time period and limited expertise on virtual conferencing - we still had an extremely informative and interactive conference.



In other exciting news, recently Keith Topper recently joined the board of directors for the foundation. Keith is our first board member to have Galactosemia.

Finally, the second round of the 2020-2022 research grant process is in full swing. There are several grant applications being reviewed and the final recipients will be announced soon.

While 2020 has been a trying year for all, the Galactosemia Foundation has tried to make the best of the hand we were dealt! We hope everyone has a great start to your 2021. Remember to fundraise for Galactosemia! We all have a great opportunity to raise awareness and funds on Rare Disease Day, which is quickly approaching on Feb. 28. Get out there and raise awareness!!

# Galactosemia Together: Applied Therapeutics launches Galactosemia Awareness Campaign



Inspired by people living with Galactosemia, developed through partnership with caregivers and loved ones, and guided by the medical community who manage this disease, GALACTOSEMIA TOGETHER was conceived!

To help connect, educate and support impacted families, Applied Therapeutics designed a comprehensive digital learning environment to address gaps in education, which includes the following key initiatives:

- **Galactosemia.com** – educational hub enhanced with rich media learning resources (<http://www.Galactosemia.com>)
- **Galactosemia Together Facebook Page** – engaging and connected learning space for Galactosemia families to meet others impacted by this disease (<http://www.Facebook.com/GalactosemiaTogether>)
- **Galactosemia Together YouTube Channel** – showcasing innovative digital 3D animation to enhance Galactosemia education (<https://bit.ly/galactosemiatogetherYT>)
  - **Galactosemia: A Tale of Two Pathways** – patient-friendly mechanism of disease video designed to support learning (<https://www.youtube.com/watch?v=QnR0GOyWhzM&feature=youtu.be>)
  - **Galactosemia: The Toxic Role of Galactitol** – HCP high-science mechanism of disease digital 3D experience to drive understanding (<https://www.youtube.com/watch?v=amXxlEbhp-Q&feature=youtu.be>)

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## Clinical Trial Update

On behalf of Applied Therapeutics, we thank the Galactosemia community for their overwhelming support and participation in the ACTION-Galactosemia clinical development program.



We plan to restart the pediatric Galactosemia trial soon after the New Year and are so appreciative of the families who have waited patiently. We believe the trial will run more smoothly because of changes implemented over the last few months.

The adult clinical trial will also begin accepting new participants into the open label extension. If you are interested in participating in either the adult or pediatric clinical trial and would like more information, please email us at [galactosemia@appliedtherapeutics.com](mailto:galactosemia@appliedtherapeutics.com).

# Volunteers Needed: A note from Dr. Judy

Once again, I am asking for your help with our research study of classic galactosemia. As always, we invite any and all individuals living with classic galactosemia to **contact me (Judy) at [jfridov@emory.edu](mailto:jfridov@emory.edu)** to learn more about our study so you can decide if you might want to enroll. Participation is non-invasive, and at no cost to you, and the more people participate, the more meaningful the study results will be for current and future patients with galactosemia.

Every volunteer is essential, and now we are also making a special effort to increase the diversity of our participant team to make sure our study results are relevant for the full rainbow of people with galactosemia. We therefore especially encourage any families with Black or African American, Native American, Asian, and/or Latinx backgrounds to join us.

## VIRTUAL CONFERENCE A SUCCESS

As COVID-19 caused many parts of the country to shut down, we contemplated canceling our semi-annual conference. One of the most important aspects of the conference is the face-to-face interactions and friendships created - how would that happen in a virtual environment? But there are other important aspects too, including education for new families, learning about on-going research and hearing directly from the doctors and researchers. With that in mind, we decided to move forward with a virtual conference. The Board made sure to incorporate networking opportunities throughout the conference. We also were able to reach many impacted families from many different countries; families who would likely not have traveled for our in person conference. See below for more stats and information from our virtual conference:


### App Activity Breakdown

 **343**  
Tickets Sold

 **292**  
Users Logged In

 **3,997**  
Profile Views

 **2,696**  
Messages

 **770**  
Bookmarks

 **144**  
Lounge Meetings

 **89**  
Medical Booth Visits

 **107**  
Corporate Booth Visits

### Participation From Around The Globe



**14 countries**

- Argentina
- Australia
- Canada
- Guatemala
- Ireland
- Israel
- New Zealand
- Norway
- Portugal
- Singapore
- Spain
- United Kingdom
- United States of America
- Undisclosed - Armed Forces Pacific

**34 US States**

- Alabama
- Alaska
- Arkansas
- California
- Colorado
- Connecticut
- Florida
- Georgia
- Illinois
- Indiana
- Iowa
- Kentucky
- Maryland
- Massachusetts
- Michigan
- Minnesota
- Mississippi
- Missouri
- Montana
- New Jersey
- New York
- North Carolina
- Ohio
- Oklahoma
- Oregon
- Pennsylvania
- South Carolina
- Tennessee
- Texas
- Utah
- Virginia
- Washington
- West Virginia
- Wisconsin



## GLOW for Galactosemia on Feb. 28

Looking for a way to advocate for Galactosemia and help us raise funds on Rare Disease Day 2021?! GLOW for Galactosemia is the perfect opportunity! GLOW is setup to be a virtual race across the U.S. where Galactosemia families can participate and invite others to show support for our Foundation!

We are thrilled to be hosting this year's GLOW on Feb 28, 2021 and looking to expand our reach into 30 states (we had 27 represented last year!).

With the help of several team leaders across the nation, we hosted 250 virtual participants last year, and raised over \$27,000 for the Galactosemia Foundation.

This year, each participant will receive a GLOW in the dark t-shirt and mask, before race day! We hope you will join us! More information can be found at [www.glowforgalactosemia.org](http://www.glowforgalactosemia.org) or email [brittany.cudzilo@galactosemia.org](mailto:brittany.cudzilo@galactosemia.org).

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## Fore the Cause: May 21

"Fore the Cause" charity golf outing will be held Friday, May 21 in Midlothian, Virginia. While Covid-19 caused the cancellation of last year's event, the Saylor family hopes to resume this annual event.

"Fore the Cause" has raised over \$300,000 for The Galactosemia Foundation since 2008 and we hope to have another successful event this year. Locally, there is a lot support from the Rodgers, Stroop and Berling families and we could not have this event without them. We are looking to add a virtual golf team option this year so stayed tuned for details! More info will be posted on Facebook and Galactosemia.org in the coming months.

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## Raising Funds and Awareness

Rare Disease Day is right around the corner. While this day is celebrated on February 29 (a rare day happening only every 4 years) we do celebrate annually on February 28. Thanks to the many families who have held fundraisers in conjunction with Rare disease day. Please consider having your own local event. Whether it is selling wristbands (email me I can ship them to you) or getting a virtual team together for "Glow for Galactosemia" it all makes an impact. Other efforts have included t-shirt sale fundraisers and Blue Jeans for Rare Jeans. Please feel free to reach out to Scott Saylor at [scott.saylor@galactosemia.org](mailto:scott.saylor@galactosemia.org) if you would like help to get started.

# Meet New Board Member: Keith Topper



My name is Keith Topper and I am from Long Island, New York. I am extremely excited to join the Galactosemia Foundation Board to continue to spread awareness of the condition that unites us all. Born in 1997 and diagnosed with Classic Galactosemia (a diagnosis that was changed to Clinical Variant Galactosemia in 2014) at five days old, I know what it's like to live with Galactosemia.

I struggled with speech apraxia as a child, and of course I've dealt with the diet my entire life. In 2019, I earned my bachelor's degree in Business Administration & Management with a concentration in Law from Boston University.

Since graduating, I have been working as a Legal Assistant at a law firm in New York City. Additionally, I am preparing to apply for law school after scoring in the top 10% of test takers on the Law School Admission Test. I enjoy spending time with my family (including my dog Mocha), exercising, and watching the Boston Celtics.

As a Board member, I look forward to continuing my passion of advocacy by working with members throughout the community to make our Foundation as beneficial as it can be for everybody.



**MARK YOUR CALENDAR:**  
**July 28 - 30, 2022**  
**2022 Galactosemia Conference**  
**Orlando, FL**

## Connect with Us

**Follow Galactosemia Foundation on Social Media and visit our website.**



## Contact The Board

**Kellie Wilcox**, President  
**Scott Saylor**, Vice President  
**Brittany Cudzilo**, Outreach Coordinator  
**Lanye Long**, Conference Committee Chair  
**Nicole Casale**, Treasurer  
**Jodie Solari**, Communications  
**Cari Miller**, Secretary