

GALACTOSEMIA GAZETTE



Galactosemia Foundation

Linked for Life

President's Message

BY: NICOLE CASALE



As we enter 2022 there are so many exciting things happening with the Galactosemia Foundation.

Conference planning is in full swing. It's hard to believe it's been four years since we've all been together, and we truly can't wait to see everyone.

A lot has been going on with the FDA and we plan to hold a Patient Focused Drug Development (PFDD) meeting with them in the near future. At this meeting, the FDA will hear from the community to learn about galactosemia and how it has affected their lives. We'll give a lot more information in the coming months, but just know your support and "virtual" attendance is vital to make our voices heard.

In other news, we have a new fundraising platform called Mobile Cause. We are so thankful to families as they raise both awareness for the disease and funds for the Foundation. This platform will make it easier to create your own fundraising event, as well as create a team as part of other events.

If you have been or are considering fundraising, you may be wondering where the money goes once the Foundation receives it. As an organization run entirely by volunteers, most of the funds go directly to support our mission and objectives: "To educate, support and provide advocacy for those affected by galactosemia as well as to network with professionals to inspire the treatment and advanced research of Galactosemia." In other words, aside from very minimal monthly and yearly expenses, a majority of donations go toward putting on conference and funding research. To date we have funded \$931,595 in research and are excited to be opening our next round of research proposals in the coming months.

So, back to conference! We can't wait to see you; registration is open, so be sure to take advantage of early bird pricing. If you are not able to make it in person, this will be a hybrid event, which means you will have a virtual option to learn and network with others. I look forward to connecting with you all!



Sea The Change!

We are only about five months away from our 2022 Conference: Sea the Change. After moving our conference virtual in 2020, we are so excited to reconnect with everyone in person. But for those of you who cannot make the in-person conference, we will have a virtual option.

The conference will provide an opportunity to learn about all aspects of galactosemia and will focus on addressing real-life situations faced by those with galactosemia. Workshops will offer practical strategies and tools to help improve quality of life for those with galactosemia and their caregivers. While adults are attending the general session, professional childcare services by trained staff will be offered for all ages. And as always, we have fun activities planned for our kids (starting at age 9) to young adults through our Generation Gs, G-Forcers and Mighty Gs programs.

You can [register today](#) through March 31 at the early bird rate, which is as follows:

- General - \$190
- Childcare - \$240
- Might G, G Force, Generation Gs - \$240
- Virtual - \$100

Hotel reservations can be made at [Renaissance Orlando at SeaWorld](#) for a special rate of \$150 plus \$25 resort fee per night, if booked by July 5. To make your reservations you can call the hotel (407-351-5555) – make sure to let them know you're attending the 2022 Galactosemia Foundation Conference. You can also book online using our group block link: [Renaissance Orlando at SeaWorld](#). For more information on rates and what the hotel has to offer, visit the [Galactosemia Foundation website](#).

Hello Orlando, Florida!

BY: *CHERI LORD*



This year's Galactosemia Conference will be in The City Beautiful or what I would consider "Where Dreams Come True."

The Renaissance Orlando at SeaWorld will be hosting this year's conference from July 28-30. If you plan on staying a few extra days before or after the conference there will be plenty to see and do! I was lucky to live in this city for over seven years and during that time I worked at both Disney World and Universal Studios Orlando. There are three major theme parks right in that area you can experience: SeaWorld, Disney and Universal

If you plan on dining inside the parks the cast members and team take diet restrictions very seriously. Before you order off the menu let your server know that you have a serious diet restriction; sometimes they will have a special menu but not always. You also can ask to have the chef come out to your table if you have questions about the menu. Usually the server can answer your questions as well. If you choose to ask the server any questions regarding the menu when they turn in the order it goes directly to the chef and they ensure it is prepared without whatever restriction you are requesting. They also make sure the plate is marked as an allergy and the food runner brings that plate out by itself. They never carry any other dishes along with the allergy plates.

If theme parks aren't your thing, you have another option...BEACHES of course!! But you must decide if you want to venture to the Atlantic Coast or the Gulf Coast. My personal favorite is the Gulf Coast on the west side of Florida. Gorgeous beaches, white sands and beautiful ocean views. The Atlantic Coast on the east side has plenty to offer with water sports and better waves. Whichever you choose you are sure to have wonderful experience!

Check out the links for the theme parks below:

[SeaWorld Theme Park - Florida Aquariums | SeaWorld Orlando](#)

[Discover Disney World Vacation Packages \(go.com\)](#)

[Get 2 Days Free with a 3-Day Ticket | Universal Orlando Resort](#)

And of course, we cannot forget what Florida is known for...GATORS. If gators are your thing, check out [Gatorland](#) for an affordable fun-filled day!

If you want help planning or booking your trip, feel free to email me at Cheri.Lord9@gmail.com and I can send you the information for travel agents who specialize in Orlando-area vacations!

Paul Pruszynski Memorial Scholarship Available

The Paul Pruszynski Memorial Scholarship was created to honor Paul's memory to provide young people with Galactosemia (GGs, G-Forcers, and Mighty Gs) who might not otherwise be able to attend the conference because of financial reasons.

Paul attended his first PGC/Generation G Conference in 2010 in Minnesota. Paul, who was 23, passed away suddenly in January 2011. Paul was from Texas, so was very much looking forward to attending his second conference in 2012 in Texas.

Generation Gs (18 years old to young adults), G-Forcers (13 to 17 years old), and Mighty Gs (9 to 12 years old) who will not be able to attend the 2022 conference (in person or virtual) because of financial difficulties can apply for the scholarship by filling out a questionnaire.

The scholarship will assist with the costs of one and possibly more of the following: hotel, registration and/or travel.

Recipients who are awarded the Scholarship will also participate in a ceremony at the conference to honor Paul's memory. If you are interested, please contact: Linda Manis: Lmscript1@aol.com



Hey G-Forcers and Mighty Gs!

We are looking forward to seeing everyone in Orlando this year. The 2020 conference was fun and everyone did an amazing job of moving the conference online but we are excited about being back in person as we do some of our normal breakouts and some new ones! The schedule is still being finalized and we are open to suggestions. Please email me at do2bb@yahoo.com if you have any questions or suggestions. Hopefully we can thaw out in Florida as we are even getting snow here in South Carolina, which is rare for us!

Silent Auction Items Needed

As always, the Silent Auction will be back with lots of great items for you to take home! The Board has been busy soliciting donations, but we need your help! We know many of you have connections, talents or items that we do not. Some ideas include:

- Homemade crafts or other items
- Theme baskets (movie night, spa, etc.)
- Tickets to an event

All proceeds go into the Foundation's general fund to help make these conferences possible! For more information contact board@galactosemia.org.

Research Study Opportunities

Calling all Infants (2 and under) with Classic Galactosemia

Researchers at Emory University in Atlanta, GA are conducting a study to define the timing, prevalence and potential changes over time of long-term complications in patients with classic galactosemia.

Participants must be younger than 2 years old at enrollment and have galactosemia. All study requirements can be completed remotely, and participants will be compensated \$50 for their first year of participation and \$25 a year for up to four additional years.

Study requirements include an online informed consent process for enrollment and then completion of a series of online surveys about your baby's medical and family history. Then every six months, through age 5 and annually after that, you will be asked to complete follow-up online surveys. Additionally, you will be asked to have a small extra blood sample drawn during annual appointments that can be returned to the study via a provided kit which will include the tube, instructions, packing materials and a prepaid shipping label. There is no cost to you or your insurance and no extra needle stick. As a benefit, babies who donate a blood sample may receive free GALT genotype analysis and girls who donate a blood sample may also receive free analyses of two hormones related to ovarian function: follicle stimulating hormone (FSH) and anti-Mullerian hormone (AMH).

More information is [available here](#). If you have questions or are interested in enrolling in the research study, contact Judith Fridovich-Keil at jfridov@emory.edu or contact the research team at galactosemia@emory.edu.

GLOW for Galactosemia on March 6

The 5th annual GLOW for Galactosemia race is set for March 6 in Knoxville, TN and virtually across the United States.

This annual fundraiser has raised over \$100,000 in the past 4 years for the Galactosemia Foundation, through sponsorships and families affected by galactosemia building teams across the US.

Virtual registration recently closed with a record breaking 41 teams! We are so excited for such a big turn out this year. Each virtual participant will receive the GLOW for Galactosemia long sleeve t-shirt (with glow in the dark ink!) + race swag, such as an Applied Therapeutics Galactosemia Together hoodie.

If you are in the Knoxville area you have until March 5 [to register and join us in person](#). We are so excited to GLOW with all of you virtually and in person on March 6!



JAGUAR

GENE THERAPY

Jaguar Gene Therapy was established in 2019 with the mission to accelerate breakthroughs in gene therapy for those living with severe genetic diseases. The team at Jaguar Gene Therapy is proud to partner with the Galactosemia Foundation to achieve shared goals. Jaguar Gene Therapy is made up of a proven and growing team of experts who have first-hand experience in bringing novel gene therapy treatments to patients and their families. Committed to patient safety and product purity, Jaguar is rapidly advancing an initial pipeline of three programs, including the JAG101 program in development for Type 1 galactosemia.

JAG101 is an investigational gene therapy that aims to deliver a gene replacement solution to address the root cause of Type 1 galactosemia by delivering the functional GALT gene via the AAV9 vector. Gene therapy offers to potentially reduce multiple toxic metabolites, bringing them closer to normal levels. The treatment is intended for younger patients, where there is urgency to potentially prevent lifelong complications. Jaguar has research agreements in place for the program with Emory University and the University of Utah, where encouraging preclinical proof-of-concept data was produced.

The JAG101 investigational gene therapy for Type 1 galactosemia is currently in preclinical (animal model) testing. The Jaguar team is working diligently to conduct the necessary preclinical studies, in compliance with all guidelines and regulations, so the JAG101 program can advance as safely as possible to human clinical trials in the coming years. There is a lot of hard work to be done, but we sincerely hope that by working together, we can help accelerate a gene therapy breakthrough for the galactosemia community. The Jaguar team is excited to join us for GLOW for Galactosemia and the 2022 annual conference.

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