

WINTER 2020  
HAPPY NEW YEAR!

# GALACTOSEMIA GAZETTE

OFFICIAL NEWSLETTER OF GALACTOSEMIA FOUNDATION



## President's Message

BY: KELLIE WILCOX, PRESIDENT

It's almost go time! And by go time I mean ...FULL SPEED AHEAD!! Which is our conference theme for the Galactosemia Conference held in North Carolina **July 16-18!** We hope everyone is ready for everything NASCAR because your Galactosemia Foundation is FULL SPEED AHEAD on planning a flag raising conference!

The conference hotel – Embassy Suites – is located in Charlotte/Concord, NC on a beautiful lush golf course.

The foundation board has been busy planning great break-out sessions to keep current on the latest changes within the industry. The talent show will be back Friday night – don't forget to sign up to reserve your spot on the race track. This event has become such a hit the schedule is truly busting at the seams and no way to expand - so sign up early!

Coordinators of the Mighty Gs, GGs and GForce have been planning like crazy for the off-site activities - read more about this in the newsletter!

The Thursday night sweet treat meet and greet is sure to please everyone this year! Again, not wanting to give away all our surprises, but you are definitely going to want to race on down to Thursday night!



Please remember this hotel has limited rooms and we want everyone to have an opportunity to stay at the conference hotel. However, there is a sister hotel close by; we will announce specifics later. We are just over 6 months til the 'RACE' for the conference starts!

We are also trying to put together a 5K Fun Run to jump start our FULL SPEED AHEAD CONFERENCE! Let us know if you would like to see this become a reality. We can't wait to see everyone soon in the great Carolinas!



## Babble Boot Camp: Clinical trial aiming to prevent speech and language struggles for infants with galactosemia

BY: DR. BEATE PETER

Children with classic galactosemia (CG) are at risk for speech and language difficulties. This has been known for decades. One would think that babies who receive the CG diagnosis shortly after birth would benefit from earliest help in the area of speech and language, but that is usually not the case. These kids experience the same assessment and treatment schedules as most other kids with speech and language problems. Delays in these areas can't be tested for until the children are old enough to show the delays, age 2, 3, or even 4 years. And that's how long many babies with CG have to wait to be tested and start treatment.

The big question is: Can speech and language problems can be prevented? In other words, can knowledge of a genetic risk for these problems be used toward earliest, proactive interventions?

The Babble Boot Camp is a clinical trial designed to test just that. The idea is simple: The newborn diagnosis of CG lets us identify those kids with the high risk for speech and language difficulties. The babies enter the study when they are very young - only 2 to 5 months old. Right from the start, they and their parents see a speech pathologist once a week. During the nearly two years that the families are in the active phase of the study, parents learn how to stimulate cooing, babble, and first words. They learn about helping to build up a nice big vocabulary and to make sentences get longer and more complex.

The children are watched very closely by the research team via monthly sound recordings of their babble and speech and also using questionnaires about general developmental milestones.

When the children are around 15 to 18 months old, the moment of truth arrives: Was the treatment effective? Are the children in the treatment group progressing faster in their speech and language development than children with CG who did not receive the treatment? If statistical testing shows this is the case, it will be the first solid evidence this method works. The team follows up with all children in the Babble Boot Camp by doing standard speech and language development testing once a year until they turn 4. If we find continuing and compelling evidence for the Babble Boot Camp's efficacy, this could be a game changer in that all children with CG should be considered for proactive treatment instead of the current wait-and-see method.

The clinical trial study is funded with a research grant from the National Institutes of Health. Principal Investigator is Dr. Beate Peter at Arizona State University. You can contact her at [Beate.Peter@asu.edu](mailto:Beate.Peter@asu.edu) if you are interested in participating. There is no cost. All materials are paid for by the study. Families receive a fee for participating in the study.

You can read more about this exciting study on the [Arizona State University news website](#).



## The Countdown to Charlotte

BY: NICK ELLIOTT and PATRICIA WATKINS

On your mark, get set...GO to Charlotte! We are excited and looking forward to seeing everyone in the Queen's City as we race to another great conference. Why is it going to be so great? Because you are going to be there, and it is going to be busy!

### GG and G Force (13-17 years)

We have a variety of things planned as we keep some of our traditions and will introduce some new things to do, based on survey results. We don't want to ruin the surprises we have planned but Charlotte is the home of NASCAR. Since many of you will be vacationing there as well, here is a list of ideas of things that we are not doing that may interest you: NASCAR Hall of Fame, Carowinds Amusement Park, Panthers Stadium, Sea Life Aquarium, National Whitewater, Music Pavilion and Daniel Stone Garden. All are within 30 minutes from Conference Center.

### Mighty Gs (9-12 years)

For the 2020 conference the Mighty Gs will have their own group and their own room! We have some fun offsite activities planned as well as some activities at the hotel. More details will be available in the June newsletter.

## Charlotte's Got A Lot: What to do in Charlotte

BY: CHERI LORD

This summer's Galactosemia Conference is in Charlotte, NC. AKA The Queen City, named after Kind George III wife Charlotte. Just a little FYI if you didn't know.

It will be the second conference for our family. My 4-year-old son Anakin has Classic Galactosemia. Denver was our first conference and it was amazing. I gained a lot of knowledge during our time there and met some great people!

While visiting the city during the Galactosemia Conference there is so much to see and do. If you are staying a few extra days and are interested in history the Reed Gold Mine State Historic Park which is almost 30 miles east of Charlotte, is a good option. This historical park - likely the birth of gold mining - is open Tuesday - Saturday 9 a.m. - 5 p.m. No fee for tours or admission into the mine but if you want to try out the gold panning the fee is \$3 per person. More information: [historicsites.nc.gov/all-sites/reed-gold-mine](http://historicsites.nc.gov/all-sites/reed-gold-mine)

Another affordable option is the North Carolina Transportation Museum. Take a scenic 25-minute train ride through the 60-acre park. More information: [nctrans.org](http://nctrans.org)

For the racing fan in your family the NASCAR Hall of Fame would be perfect! Lightning McQueen is even on display! Plus, an iRacing simulator. More information: [nascarhall.com](http://nascarhall.com)

Now, my favorite part is food! Anytime I visit a new place it's always fun to try some fun and exciting new restaurants and Charlotte's got a lot to offer. For an organic plant-based dining experience, Living Kitchen is one option in the area. They have a juice menu and a cashew-based cheese plate! More information: [livingkitchen.com](http://livingkitchen.com)

There is also The Cowfish - a sushi burger bar. I have eaten at the one in City Walk at Universal Orlando Resort and love the experience.

Visit [charlottesgot alot.com](http://charlottesgot alot.com) for more to do in this fun city!





## Ready, Set, GLOW

BY: BRITTANY CUDZILO

It's almost GLOW time once again! And this year we get to share a weekend with Rare Disease Day, making it more special and exciting to pull in new faces and community as we work together to raise awareness, funds, and build up a stronger Galactosemia community.

In its second year (2019), GLOW for Galactosemia had participation from over 20 states. And out of its 350 sign ups, 150 were virtual - meaning the participants were from somewhere outside of the main event in Knoxville, TN. The event raised over \$20,000 that went straight into the general fund for the Galactosemia Foundation, and so much of that was from our Galactosemia community sharing the opportunity to be part of this event and to give! One of the best parts of this event is it gives us a way to work together, even when there are many miles that separate us.

How can you get involved? There are two big ways, each one crucial to the success of the event! The first way is signing up and building a team where you are! It's very easy - you can visit [runsignup.com/Race/TN/Knoxville/GLOWforGalactosemia2019](https://runsignup.com/Race/TN/Knoxville/GLOWforGalactosemia2019) and set up your virtual team, share the opportunity to get involved with your family and friends (maybe sharing your personal story with Galactosemia, and/or why this is something you want to be part of), and pick a spot to meet and get active! You do not have to walk or run, though many participants do either at an indoor track or local park. We also have had some groups participate in a spin class with a donated space and instructor! Get creative! Each person who signs up on the website will be sent a t-shirt (SUPER comfy and glows in the dark) and other fun event goodies!

The second way we need your help is sponsorship! You don't have to be close to the main event in Tennessee to be a sponsor. We sent shirts all over the US last year and had several sponsors who were not local. If you have a connection with a business, we would love to speak with them about partnering with us to raise funds for the foundation. We offer logos on the back of the shirt, website recognition and personalized social media shout outs...and it's a tax-deductible donation, meaning we are an IRS charitable foundation.

Sponsorship is due by Feb. 1 so t-shirt designs can be completed for printing, and each virtual participant must be signed up by Feb. 14 to receive their packets before GLOW day. This seems far away, but by the time we have another newsletter we will be celebrating all that happened!

We are REALLY excited about what this year will bring for GLOW and for our foundation. We would LOVE to partner with you to make this year memorable! Please feel free to reach out to [brittany.cudzilo@galactosemia.org](mailto:brittany.cudzilo@galactosemia.org) with questions, comments, and/or ways to partner with you or others!

# PAUL PRUSZYNSKI MEMORIAL SCHOLARSHIP

BY: LINDA MANIS

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

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 he was very much looking forward to attending his second conference in 2012 in Texas.

Generation Gs (18 to young adults), G-Forcers (13 to 17), and Mighty Gs (9 to 12) who cannot attend the 2020 conference 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 would like to be considered for the scholarship, please contact Linda Manis: [Lmscript1@aol.com](mailto:Lmscript1@aol.com). If you would like to donate to the memorial fund [click here](#).**

## 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! All proceeds go into the foundation general fund to help make these conferences possible! For more information contact [brittany.cudzilo@galactosemia.org](mailto:brittany.cudzilo@galactosemia.org).



## Connect with Us

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## Contact The Board

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