

GALACTOSEMIA GAZETTE

OFFICIAL NEWSLETTER OF GALACTOSEMIA FOUNDATION



NORD Rare Disease and Orphan Products Breakthrough Summit

BY KELLIE WILCOX, PRESIDENT

As a representative of the Galactosemia Foundation, I attended the NORD Rare Diseases and Orphan Products Breakthrough Summit in Washington, DC this fall. NORD is the National Organization for Rare Disorders, and the Galactosemia Foundation has been a patient organization of NORD's for several years. NORD provides the foundation a level of advocacy we are not capable of on our own and also helps the foundation with promoting our cause through social networking, educational opportunities and research development.

The summit pre-meeting was a very stimulating Patient Organization Membership Meeting. I met several leaders from other rare disorder organizations. We were given the opportunity to have a liaison from NORD and collaborate with each other. We learned about best practices, shared ideas that could be used within similar organizations and discussed common hurdles that affect all our organizations.

On the first day of the summit, we heard from a few of the ambassadors of NORD who are focused on advocacy. They talked about the importance of designating someone within your organization who can commit their time to grassroots advocacy.

That afternoon was a breakout session on the challenges for orphan products, in which we heard a lot about the importance of a [patient registry](#).

Our "foot in the door" (so to speak) for advancement in drug therapy, recognition or increased research starts with ensuring our patient registry is complete and functional. They have to know who we are and what we are about! If you have not joined our patient registry, I encourage you to [do so here](#).

On Day 2, we heard details from case studies of gene therapy and clinical trial successes. I learned a lot about what goes into actually getting to the point of opening the door to the trial. One of the most interesting things about Day 2 was the FDA's comments and learning how the process works. I, now, better understand some of the background requirements. The FDA is opening a new patient outreach program and is very interested in the different areas of rare disease and where their current clinical trials are headed.

The summit was an eye-opener for me in many areas. I am very grateful for the opportunity to have attended. I encourage everyone to learn more about what NORD can do for our foundation and you personally!



RESEARCH:

Metabolic Perturbations in Classic Galactosemia Beyond the Leloir Pathway: Insights from an Untargeted Metabolomic Study

BY: S. TAYLOR FISCHER
ALLISON B. FREDERICK
VILINH TRAN SHUZHAO
LI DEAN P. JONES
JUDITH L. FRIDOVICH-KEIL

Dr. Judy shared the following research with volunteers with a note that said, "Of course, none of this work would have been possible without the Galactosemia Foundation and all the wonderful volunteers who participate in our research, so thank you!"

Published in Journal of Inherited Metabolic Disease
(January 2019) <https://doi.org/10.1002/jimd.12007>

SUMMARY

What was the goal of this study?

The goal of this study was to explore mechanism in classic galactosemia (CG). Specifically, we wanted to define the larger-scale metabolic consequences of GALT-deficiency in children and adults with CG.

Rationale

Many prior studies have looked at metabolites related to galactose metabolism in people with CG and controls, reporting, for example, that galactose, galactose-1-phosphate, and galactitol all tend to be elevated in people with CG, especially infants exposed to dairy. But are these the only differences? We wanted to look at metabolites outside the set traditionally tested to see if this approach might shed light on other underlying metabolic pathways that are perturbed in CG. We also wanted to test whether some of these metabolites might distinguish people with CG who experience a more severe long-term outcome from people with CG who experience a milder long-term outcome.

What did we do?

We used a method called liquid chromatography-mass spectrometry (LC-MS) to separate and measure the abundance of thousands of small molecules (features) in samples of plasma from 183 people with CG and 31 controls.

Using statistics, we then compared the patterns of features detected in samples from people with CG versus controls, and also between people with CG who experienced mild versus severe outcomes. Next, we mapped as many of the features as possible that distinguished people with CG from controls to known metabolites based on properties observed in the LC-MS. Finally, we mapped as many of these metabolites as possible to recognized biochemical pathways.

What did we learn?

We learned that there are hundreds of small molecules that show significantly different abundance in the plasma of people with CG compared to controls. Some molecules are found at unusually high levels in people with CG, and others are found at unusually low levels in people with CG. These molecules mapped to many different biochemical pathways including those related to oxidative stress, amino acid metabolism, and mitochondrial function. One feature, that unfortunately could not be mapped to a known metabolite, also distinguished girls and women with CG who have premature ovarian insufficiency from girls and women with CG who show evidence of milder ovarian outcome.

Why is this important?

The results of this study confirm that CG impacts many biochemical pathways beyond galactose metabolism and offer a new approach to identifying novel targets for intervention.

Research Team Recognizes Christy Johnson for 15 Years of Service

BY: RESEARCH TEAM

Thanks to Christy Johnson for her 15 years of service on the Galactosemia Foundation Research Team.

Christy Johnson, past chair of the Galactosemia Foundation Research Team, has been a member of the Research Team since its inception as the Parents of Galactosemic Children (PGC) Research Committee in 2004. She was instrumental in developing the research grant program, working primarily with Thérèse Cozzo, the first chair of the PGC's research committee, and Jo Beth Southard.

Thérèse recalls:

"I am not only appreciative for Christy's years of service to the Research Committee, but also for helping us many years ago in the early stages to get the Research Committee up and running. Christy was a wonderful source of support, positivity and knowledge. It was a pleasure to work with her. I will be forever grateful for all that she has done for this committee and the organization. Thank you, Christy!"

"Christy supported the research committee through two rounds of grants, where PGC awarded three grants. This was an exciting time for the committee as many in the PGC were raising money for the research fund and we were relieved when a handful of researchers actually submitted proposals to help our kids."

Jo Beth recalls:

"Christy was instrumental in the development and organization of our Research Team. She was always positive, and I always respected her opinion and really enjoyed working with her on the team. When we needed someone to step up and take a role or a duty, she was always there. The Galactosemia Foundation was lucky to have her, and she leaves big shoes to fill. Thank you, Christy, for all you have done!"

"Christy led the research committee in Round 3, which led to the awarding of three additional grants. Funding included research on the galactosemic diet and the first grant awarded outside the United States. The success of the research program during this time encouraged the board to rename PGC the Galactosemia Foundation (GF). And the research committee became the research team."



Dan Lambert remembers:

"Christy was a great leader of the research team. Since the research program was really growing, she had to recruit the additional professionals needed to peer review the proposals and recruited new people to join the research team. She communicated well with the PGC leadership on the board. I don't know what we would have done without Christy in those days."

"Christy has continued supporting the research team through four additional rounds (eight years), leading to the awarding of 11 additional grants. She has served as the wise resource for team members as they come and go on the research team. She has helped the Galactosemia Foundation move into new areas, like the development of a Galactosemia Patient Registry."

Kelley Foley writes:

"I will be forever grateful for Christy. She has taught me so much, and the program truly would not be where it is without her efforts. I don't think people really recognize how much of her time and expertise she has dedicated to our community. She has led with great patience, kindness and wisdom. She has always made herself available to answer questions, guide us and get things done. Christy could never be replaced and will be extremely missed. She is a person of great integrity and has truly been a mentor for me. I have enjoyed working with her over the last several years. Thank you, Christy, for the many years of service!"





Galactosemia

Galactosemia Patient Registry Needs You

BY: JODIE SOLARI

We all have different experiences with Galactosemia – from having a child dealing with many challenges to a child who experiences few issues to being an individual with Galactosemia. But I think we all agree if we could do something to make the future easier and better for those with this disease, we would.

Did you know you can? And it is as simple as registering your child (or yourself for those with Galactosemia) in the Galactosemia Patient Registry. The registry is an online data system that collects, stores and retrieves patient data for research studies. The purpose of the registry is to:

- Assist researchers studying Galactosemia and facilitate recruitment for research studies;
- Characterize and describe the Galactosemia population;
- Help with the development of recommendations for standards of care;
- Conduct a study that will improve the understanding of Galactosemia and its progression over time.

Because Galactosemia is so rare, it is important to add as many people as possible to the patient registry to gather accurate and complete data. Participants are encouraged to update their information every two to three years through adulthood to document any changes in the progression of the disease.

The registry follows strict government guidelines to assure patient information is protected. It collects the following information:

- Socio-demographics;
- Medical and diagnostics;
- Treatment and disease progression;
- Management of care;
- Quality of life.

The Galactosemia Patient Registry can be accessed on the homepage of [the Galactosemia Foundation's website](#).



2020 Conference Planning Underway

BY: KELLIE WILCOX

We hope everyone is getting excited about the 2020 Galactosemia Conference in North Carolina from July 16-18. The board of directors and conference committee are busy planning a great conference in a fun, busy town! We have listened to your feedback from the 2018 Denver Conference and are making changes.

Since we're starting to plan, we thought maybe you are, too. So we wanted to share some information to get you started! The conference will be held at Embassy Suites by Hilton Charlotte/Concord Golf Resort & Spa in Concord, North Carolina, about 25 miles northeast of Charlotte. Concord is a small suburb and has several activities, shopping venues and restaurants to keep you busy. The airport is about a 30 minute drive, so plan accordingly if you are flying in.

We are very excited about the hotel. It has a few amenities our conference hotels have not provided in the past, including a hot continental breakfast and a pull out sofa in every suite! That right there is a dream for families!

The conference committee will work with the hotel chefs to have safe foods on the breakfast buffet. BUT the hotel also has a made-to-order omelet bar - the line will be long, so plan ahead! In addition to a sofa in every room, there is a mini refrigerator, microwave and coffee pot. With amenities like these and less than 300 double-occupancy rooms, this hotel is going to book fast, so do not delay when registration opens in early 2020. There is a sister hotel nearby for any late registrants.

All activities including breakouts, childcare and silent auction, etc., will be located on the same floor, making transitions easy between events. We are excited to announce the return of the Mighty G program, which hasn't been active in several years! This is a great group comprised of kids who are too old for childcare but too young for the older group and their activities.

Also...while we haven't announced the theme yet, Charlotte is the home of NASCAR. These are just a few highlights of the 2020 Conference in North Carolina, and we hope you are getting as excited as we are! Watch Facebook, our website and the newsletter for more exciting details!



Fundraising Update

BY: SCOTT SAYLOR

WOW! I can hardly believe it has been almost a year since we last met in Denver. I am excited to see everyone in Charlotte in 2020!

Speaking of conference, it is never too early to start planning. The board has already begun planning and we **NEED YOUR HELP!** We are looking for raffle and auction donations for the conference. Unique items sell the best, but anything helps. Also, we welcome families to put together theme baskets (e.g., movie night). If you know a business that would like to support our conference or donate an auction item, please contact a board member.

"Glow for Galactosemia" ran by the Cudzilo family had their second event and raised more than \$20,000! This event is growing and is one to watch! Read the Glow for Galactosemia update for the details.

"Fore the Cause" - my fundraiser in Richmond, Virginia - is taking 2019 off. We expect to be back in 2020. A lot going on personally led to this somber decision. We have done this charity golf outing for 10 years and have raised more than \$300,000 and needed a break to attend to personal matters.

Conference Incentive will be similar to 2018. Information will be updated on the website soon. All donations to the General Fund count toward the incentive program, so start raising funds now!

And I will end with a quote attributed to Denzel Washington: "At the end of the day, it's not about what you have or even what you've accomplished. It's about what you've done with those accomplishments. It's about who you've lifted up, who you've made better. It's about what you've given back."

Ever wondered how to support our foundation?

- Set up a monthly or annual family donation.
- Check to see if your company matches donations.
- Conduct a fundraiser (walk, run, golf, party, etc.)

Reach out to me or any board member if you need help.

GLOW for Galactosemia

BY: BRITTANY CUDZILO



GLOW for Galactosemia was a HUGE success this year for many reasons! We expanded our virtual run to include more than 150 participants from 22 states which brought in more of our community to help fundraising and advocacy efforts! Our largest team was 60+ in Pierre, South Dakota and we had several teams that reached ten or more people.

What a joy it is to bring all of us together with the common goal of supporting our foundation, and support one another in our hope to fund more research and encourage more outreach.

The entire event raised more than \$21,000 for our foundation, and was only possible because of the support of our Galactosemia community. Thank you to everyone who participated! We are looking forward to making GLOW 2020 a truly special event as it will be held the same weekend as Rare Disease Day on March 1!



Follow [Glow for Galactosemia](#) on Facebook for updates. Please email brittany.cudzilo@galactosemia.org with any questions or interest in participating in 2020!

GALACTOSEMIA FOUNDATION

WELCOMES NEW BOARD MEMBERS



Brittany Cudzilo, Outreach Coordinator

Brittany is the mother of three daughters, the youngest of whom has Classic Galactosemia. Brittany and her husband, Ben, are located in Knoxville, Tennessee, and have been making connections with those around the area since their daughter, Ansell, was born. They are committed to reading the latest research and have a desire to understand Galactosemia to their greatest ability.

Brittany's background is in secondary education, and she taught high school for five years. Since leaving, she has been at home raising their girls and is now working to build the GLOW fundraiser to connect Galactosemia families and raise awareness and support for the Galactosemia Foundation. Brittany will serve as the foundation's outreach coordinator and is excited to take on this new role.

Brittany and Ben traveled to Denver for their first Galactosemia Foundation conference in July 2018 and look forward to bringing their girls to Charlotte in 2020.



Jodie Solari, Communications Lead

Jodie Solari lives in Williamston, Michigan with her husband Chris. They have three daughters, Francesca and twins Amelia and Annabelle. Francesca and Amelia have Classic Galactosemia. Soon after Francesca was born, Jodie knew she was going to be a mom with a cause and is passionate about educating others about Galactosemia. She first became involved with the foundation after attending the Atlanta conference in 2016.

Jodie is a Communications Consultant for a utility company, where she coordinates, creates and distributes internal communications to more than 10,000 employees and contractors across the state. She manages the company's intranet, monthly electronic newsletter and helps internal clients deliver the right message to the right audience at the right time. Jodie is looking forward to using her knowledge and skills as the Galactosemia Foundation's Communications Lead.



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
Karen Greenberg, Board Member at Large