

## GALACTOSEMIA GAZETTE



# Galactosemia Foundation

*Linked for Life*

**THE WAIT IS OVER - CONFERENCE REGISTRATION IS OPEN!**



We appreciate the community's patience as we worked through some technical issues with getting conference registration launched. We are excited to announce that registration is now available and the Early Bird rate will be extended until May 30. We are taking payment through PayPal. If you do not have a PayPal account you can enter your credit card information for payment. We are still experiencing some issues with registration. If you are unable to register or have any issues please email [Outreach@galactosemia.org](mailto:Outreach@galactosemia.org).

[Register Now](#)

[Book Your Stay](#)

We can't wait to see you; registration is open, so be sure to take advantage of early bird pricing. We will be adding information to our [website](#) so check there for updates.

# Paul Pruszynski Memorial Scholarship Fund

We are still accepting applications for the 2024 Conference Paul P. Memorial Scholarship Fund.

Established in honor of Paul Pruszynski, the scholarship is designed to provide financial assistance to help individuals with galactosemia attend the Galactosemia Foundation Conference. Scholarship funds may be awarded to individuals who meet certain criteria. Visit our website to learn more and apply for scholarship funds. The application period will close on May 31, or when the allocated funds are depleted, whichever occurs first. Applications will be reviewed on a rolling basis. Applicants will be notified of the status of their application by June 15.



**[Learn more and apply here](#)**

## Silent Auction Items Needed

The Silent Auction will be back with lots of great items for you to take home. But we need your help! We know many of you have connections, talents or items that we do not. Would you consider donating something to our silent auction?

Some ideas include:

- Homemade crafts or other items
- Theme baskets (movie night, spa, etc.)
- Tickets to an event
- A vacation home or Air bnb property
- Electronics
- Sports Memorabilia
- Collectables
- Unique Art/Painting



All proceeds go into the Galactosemia Foundation's general fund to help make these conferences possible! For more information contact [board@galactosemia.org](mailto:board@galactosemia.org).

**[Fill out this form to donate](#)**

# Schedule

Thursday, July 18, 2024		
Time	Session Description	Speaker
2 - 8 PM	On-Site Research: Please participate in the on-site research being offered!	
3:30 - 4 PM	Volunteer Orientation	
4:30 - 6 PM	Galactosemia 101	Brittany Cudzilo; Nicole Casale; Maureen Bell
6 - 8 PM	General Registration	
6 - 8 PM	Generation-G; G-Force Registration & Activity	
6 - 8 PM	Thursday Night Social	
6 - 8 PM	Advocacy Meet & Greet & Letter Writing	Gillian Sapia
6:30 - 7 PM	Research Committee Meet and Greet	Kelley Foley
Friday, July 19, 2024		
Time	Session Description	Speaker
8 - 8:30 AM	Any late registration	
8:30 - 9 AM	Childcare Dropoff	
7 AM - 7 PM	On-Site Research: Please participate in the on-site research being offered!	
9 - 9:20 AM	Welcome to the 2024 Conference	Nicole Casale
9:20 - 9:40 AM	Fireside chat -Panel of speakers with Galactosemia	Various Community Members
9:40 - 10 AM	Meet the board- what we do and how to get involved!	Board Members
10 - 10:15 AM	Treasurer's Report; Fundraising; Recognition and announce Incentive Winners	Scott Saylor
10:15 - 10:40 AM	Research Team- Give Updates	Kelley Foley
10:40 - 11:10 AM	Applied Therapeutics Update	Dr. Shoshana Shendelman
11:10 - 11:20 AM	Close of General Session	Brittany Cudzilo
11:20 AM - 12 PM	Roundtable- Meet and Greet	
12 -1:10 PM	Galactosemia Foundation sponsored lunch @12:00	
1:15 -2:15 PM	Breakout Series #1	Various Speakers
2:15 - 2:30 PM	Travel between breakout sessions	
2:30 - 3:30 PM	Breakout Series #2	Various Speakers
3:30 - 3:40PM	Travel to General Session	
3:40 - 3:50PM	General Session- Welcome Back	Board Member
3:50 -4:20PM	Funded Research - Back to Our Roots: Harnessing Power of Natural Remedies	Dr. Balakrishnan
4:25 - 4:55PM	Funded Research- Adult Study	Dr. Berry; Dr. Judy; Dr. Estelle
4:55 - 5:05 PM	Closing---Day Recap/Announcements	Nicole Casale
5:15 - 5:30 PM	Pick up children from childcare	
5:15 - 6 PM	Designated meetup space for families that want to connect	
5:15 - 7:30 PM	Talent Show Rehearsal	
7:30 - 9 PM	A Grand Night for Talent Together evening social	

# Schedule

Saturday, July 20, 2024		
Time	Session Description	Speaker
6 - 9 AM	Breakfast on your own	
8:30 - 9 AM	Childcare drop-off	
7 AM - 7 PM	On-Site Research: Please participate in the on-site research being offered!	
9 - 9:15 AM	General Session- opening remarks	GF Board Member
9:15 - 9:50 AM	Funded Research- Brain connectivity and metabolomics underlying cognition and motor functions	Dr. Gautschi
9:50 - 10:25 AM	Funded Research- "Is Cellular Senescence Relevant for Galactosemia?"	Dr. Masuda
10:25 - 10:30 AM	Close of Morning General Session	Board Member
10:30 - 10:45 AM	Travel to breakout sessions	
10:45 - 11:45 AM	Breakout Session Series #3	Various Speakers
12 - 1:15 PM	Galactosemia Foundation sponsored lunch @12:00	
1:15 - 2:15 PM	Breakout Session Series #4	Various Speakers
2:15 - 2:25 PM	Travel to final breakout	
2:25 - 3:25 PM	Breakout Session Series #5	Various Speakers
3:25 - 3:35 PM	Travel to final General Session	
3:35 - 4:05 PM	Welcome to our Final General Session	Brittany Cudzilo & Jodie Solari
	Galactosemia Angels--We will remember!	
	Paul Pruszynski Scholarship award	
4:05 - 4:50 PM	Panel of Doctors Q&A	Various Speakers
4:50 - 5:05 PM	Conference Closing Remarks & 2026 Conference location Announcement	Nicole Casale
5 - 5:30 PM	Pick up children from childcare	
5:15 PM - 6 PM	Designated meetup space for families who want to connect	
7:30 - 10:30PM	Saturday Night Social and Dance	

# Breakout Schedule

## **Breakout 1: Friday 1:15 to 2:15 PM**

- What to Expect in first 9 years (Dr. Gerard Berry)
- Whole-body Galactose Oxidation as a Robust Functional Assay to Assess the Efficacy of Gene-Based Therapies in a Mouse Model of Galactosemia (Dr. Bijina Balakrishnan, Ph.D)
- Long-term Outcomes in Galactosemia (Dr. Judy Fridovich-Keil, Ph.D)
- Fertility and Fertility Preservation in Classic Galactosemia (Dr. Veronica Gomez-Lobo)
- Rare Disease Advocacy (Gillian Sapia, RN)
- Voices of Galactosemia - Caregiver Perspectives (Applied Therapeutics)

## **Breakout 2: Friday 2:30-3:30 PM**

- Treating Speech and Language Disorders in Galactosemia (Dr. Nancy Potter & Victoria Heinlen)
- Tentative: Gene Therapy
- Updates from the Fridovich-Keil Lab & Gene Therapy and Bone Marrow Transplant Data (Dr. Judith Fridovich-Keil, Ph.D)
- Mental Health and Galactosemia (Dr. Goodlett)
- Bones Basics, Calcium, Vitamin D & Beyond (Sara Erickson & Mackenzie Lane)
- Managing Pharmacy issues for Patients with Galactosemia (William Bell)

## **Saturday**

### **Breakout 3: Saturday 10:45-11:45 AM**

- Experimental GALT mRNA Therapy Improves Motor Performance of a Mouse Model of Classic Galactosemia (Kent Lia)
- Molecular Fingerprints at Different Developmental Stages in a Galactosemia Zebrafish Model: What do they tell us? (Dr. Estela Rubio-Gozalbo & Naomi Vos)
- Conversations with you Galactosemia Children (Natalie & Anna Whittington)
- Transition & Identity Achievement for Teens and Adults with Galactosemia (Dr. Goodlett)
- Nutrition Management for Galactosemia in Infants & Toddlers (Sara Erickson & Mackenzie Lane)
- Advocating for your Child: Educational Planning for Children with Galactosemia (Tracey Miller)

### **Break out 4: Saturday 1:15-2:15 PM**

- Babble Boot Camp Team Early Intervention for Infants and Toddlers (Dr. Nancy Potter & Victoria Heinlen)
- Is Cellular Senescence Relevant for Galactosemia? (Dr. Masuda)
- Brain Connectivity and Metabolomics Underlying Cognition and Motor Functions (Dr. Gautschi)
- Behavior, Self-Management and Strategies (Dr. Dorothy Zhang)
- Nutrition Management for Galactosemia in School Age & Beyond (Sara Erickson & Mackenize Lane)
- Fundraising 101: How to host a fundraiser and where does the money go when donated to the Galactosemia Foundation? (Brittany Cudzilo & Scott Saylor)

### **Breakout 5: Saturday 2:25-3:25 PM**

- Applied Therapeutics: Modern Understanding of Galactosemia
- Jaguar Gene Therapy: Insurance Claims Analysis on the Burden of Disease Among U.S. Galactosemia Patients



# GOT TALENT?

Don't miss this amazing opportunity to show off your skills at this summer's conference. We are looking for dancers, singers, instrumentalists, comedy....you name it! This event is for all ages and stages, and it's something our conference goers have looked forward to every time.

Let's face it...our journeys are not always easy. So, this is one big way we can focus on the positives! Sign-up through the conference [registration form](#), and the sign-up deadline will be July 18. Not sure if you or your child is brave enough for a solo act? No worries....we will have several group acts where they can have fun alongside their peers. Group Rehearsals will be during the day on Friday with the Dress Rehearsal and Performance on Friday evening.

**Sign up today!**

Still have questions? Reach out to Natalie Whittington at 334-220-3092 or [natalie.a.whittington@gmail.com](mailto:natalie.a.whittington@gmail.com) for more information.

## **We Want your Feedback!**

If you were at our last conference you most likely received a copy of *Navigating Galactosemia Life Stages: A Handbook for the Galactosemia Community*, a resource from the Galactosemia Foundation with support from Jaguar Gene Therapy. If you didn't attend conference you may have seen the link in our social posts or on our website.

Navigating Galactosemia Life Stages: A Handbook for the Galactosemia Community presents information for each life stage of galactosemia to guide patients and caregivers from newborn diagnosis through adulthood as they navigate the lifelong complications of the disease.

We are now seeking your feedback on the handbook, and we hope you can fill out [this brief survey](#).

### **Navigating Galactosemia Life Stages**

A Handbook for the Galactosemia Community



2022 Edition



## What to Expect

The Galactosemia 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. 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 May 30 at the early bird rate, which is as follows:

- General - \$190
- General +offsite - \$240
- Childcare - \$240
- Might G, G Force, Generation Gs - \$240

Hotel reservations can be made at Embassy Suites by Hilton Charlotte Concord Golf Resort & Spa.

New this year we are offering a “General +off-site,” registration. This is for individuals 18+ who are either an individual w/galactosemia, or are a spouse, parent, grandparent, significant other, etc., who want to attend offsite activities with an Age Group attendee. They will miss portions of general sessions and/or breakout sessions.

Learn more about the off-site activities:

- Friday - Charlotte Motor Speedway
- Saturday - Frankie's

## 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