

GALACTOSEMIA GAZETTE



Galactosemia Foundation

Linked for Life

President's Message

BY: NICOLE CASALE



Hope everyone is doing well, there is so much going on with the Foundation now and in the coming months. For now I will only speak on conference, but don't forget about the EI-P.F.D.D. on Thursday Sept. 1, 2022 (more information to come).

I'm sure everyone is aware, but CONFERENCE! It's here and I'm hoping to see you, whether in person or virtually. Can you believe the last time we were together was Denver? We have lots of great speakers, topics, and for the first year, if you are unable to attend in person, there is a virtual option for general session and breakouts.

There are so many things that my family and I are looking forward to. Seeing old friends, making new ones, the sweet treat buffet on Thursday makes the list too. And who doesn't love Florida in July? And lastly food! We are looking forward to having safe food to order, how often do you get to go to a hotel and just choose things off the menu without worry!? Probably not very often! We'll have the Foundation sponsored lunch on Friday and Saturday in the Crystal Ballroom. Breakfast will also be sponsored by the Foundation on Friday morning only.



THE COUNTDOWN TO CONFERENCE...

Here's what you need to know!

CONFERENCE AND HOTEL INFORMATION

Registration is still open for the conference, [click here](#) to register today.

General - \$250

Childcare - \$300

Might G, G Force, Generation Gs - \$300

Virtual - \$100

Hotel reservations can be made at Renaissance Orlando at SeaWorld. 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](#).

We are excited to announce that we will be offering a complimentary breakfast on Friday morning!

WHOVA: 2022 GALACTOSEMIA FOUNDATION CONFERENCE OFFICIAL EVENT APP

Download the Whova app for information about the conference and to connect with other attendees. Attendees must register for the conference before downloading the app. The event invitation code is Galactosemia2022

With the app you can:

- Explore the professional profiles of event speakers and attendees
- Send in-app messages and exchange contact info
- Network and find attendees with common affiliations, education, shared networks and social profiles
- Receive update notifications from organizers
- Access the event agenda, GPS guidance, maps and parking directions

Sign up for Friday Night's Talent Together

We are looking forward to gathering on Friday night for our Talent Together event. Do you have a hidden, or not so hidden, talent? We would love to see it. Calling all singers, dancers, magicians, poets, jugglers, instrumentalists, jokesters and actors! Your deadline to register is July 15. Dress Rehearsal will be Friday evening 5:30-7:30 p.m. with the event from 7:30-9 p.m. All acts should be 4 minutes or under and feature music, lyrics, dress and acts appropriate for a family audience. To register fill out this form. For more info or any questions, please email TalentTogether@galactosemia.org.



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
- Vacation home or Air B&B rental

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



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SILENT AUCTION



CAPE COD VACATION!

OFFER: One-week Spring or Fall stay in a charming, historic home in Orleans, Massachusetts. Relax on your own private beach, just a 1/2 mile walk or drive from the house. Play croquet in the secluded yard, nap on the hammock, eat on the deck. House sleeps up to six in three bedrooms (twin bunk bed, double bed, and king bed) and has one full bathroom plus an enclosed outdoor shower. Well-equipped with washer/dryer, dishwasher, charcoal grill, TV, Wi-Fi, lots of books and puzzles.

(Past Galactosemia Foundation families have vacationed in the house while in Massachusetts to see Dr. Berry at Boston Children's Hospital)

VALUE: \$1,500
STARTING BID: \$400

DETAILS: 1-week stay (Saturday to Saturday) in September 2022, June 2023, or September 2023. For current availability and more information, see <https://www.vrbo.com/1664537> or contact Christy Johnson (919-357-7345 / cmjcmj0@gmail.com)

RESTRICTIONS: No pets allowed; Transportation not included to/from Orleans, MA; linens not provided (rental suggestions available)

Specially Priced Disney Meeting/Convention Theme Park Tickets Available



Are you planning to visit any Disney properties while you're in town? If so, we've got great news – because we are holding our conference in Orlando we are eligible for discounted tickets. If you have already purchased your tickets, call the number below to have the discount applied.

To purchase specially priced Disney Meeting/Convention Theme Park tickets, please call 407-566-5600 or [CLICK HERE](#). Tickets are valid 7 days pre, during, and 7 days post meeting dates. The cut-off date for advance purchase savings 7/27/22. With the new Disney Theme Park Reservation System and park capacity limitations, it's important to lock in your visit as soon as possible. A Park Reservation and ticket are required for entry.

Here are some other helpful links to help you reserve your tickets:

- Park Reservation Availability: <https://disneyworld.disney.go.com/availability-calendar/>
- Full instructions can be found here: <https://disneyworld.disney.go.com/experience-updates/park-reservations/>
- Park Hours: <https://disneyworld.disney.go.com/calendars/month/>

Connect with Us

Follow Galactosemia Foundation on Social Media and visit our website.



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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
Kelley Foley, Board Member at Large

Conference



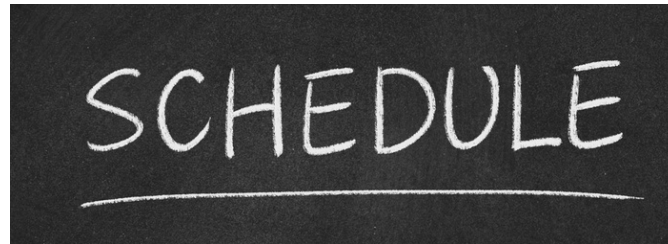
THURSDAY, JULY 28

- 9 a.m. - Yoga/Networking Event on R-Lawn
- 2-8 p.m. - On-Site Research with Dr. Judy, Dr. Berry & Dr. Rubio-Gozalbo
- 3-8 p.m. - Generation-G and G-Force Registration
- 4:30-6 p.m. - Galactosemia 101 with Brittany Cudzillo, Nicole Casale and Maureen Bell
- 6-8 p.m. - General Registration
- 6-8 p.m. - Sweet Treat Buffet
- 6:30 - 7:30 p.m. - Research Committee Meet and Greet with Kelly Foley

FRIDAY, JULY 29

- 7:30-8:30 a.m.- Galactosemia Foundation Sponsored Breakfast
- 8:30-9 a.m.- Childcare drop-off, Late Registration for GG and GForce
- 7 a.m.-7 p.m.- On-Site Research with Dr. Judy, Dr. Berry & Dr. Rubio-Gozalbo
- 9-9:20 a.m.- Welcome to the 2022 Conference with Ben & Nicole
- 9:20-9:30 a.m.- Key Note: Speaker with Galactosemia - D.J. Trainor & Dr. Nancy Potter
- 9:35-9:50 a.m.- Meet the GF Board - what we do and why you should join us!
- 10-10:20 a.m.- Fundraising; Mobile Cause; Recognition and announce incentive winners
- 10:30-10:55 a.m. - FDA - PFDD with Debbie Drell
- 10:55-11:05 a.m.- NORD - Patient Registry with Nicole Casale
- 11:05-11:30 a.m.- Research Team Updates with Kelley Foley
- 11:30-11:50 a.m.- Applied Therapeutics Update with Dr. Shoshana Shendelman
- 11:50 a.m.-12 p.m.- Close of General Session with Ben Cudzillo
- 12-1:30 p.m.- Galactosemia Foundation Sponsored Lunch
- 1:30-2:30 p.m.- Breakout Series 1
- 2:30-2:45 p.m.- Travel to next session
- 2:45-3:45 p.m.- Breakout Series 2
- 3:45-4 p.m.- Travel to General Session
- 4-4:05 p.m.- Welcome Back
- 4:05-4:35 p.m.- Funded Research: Marching Towards a cure for CG with Dr. Lai
- 4:35-5:05 p.m.- Funded Research: Back to our roots: harnessing the power of natural remedies to treat CG with Dr. Hagen-Lillevik
- 5:05-5:15 p.m. - Closing -- Recap/Announcements with Nicole Casale
- 5:15-6 p.m.- Pick up children from childcare
- 5:15 - 6 p.m.- Designated meetup time and space for families to connect
- 5:15-7:30 p.m.- Dinner on your own
- 7:30-9 p.m.- A Grand Night for Talent Together evening social

Conference



SATURDAY, JULY 30

6-9 a.m.- Breakfast on your own

8:30-9 a.m.- Childcare drop-off, Morning Coffee

7 a.m.-7 p.m.- On-Site Research with Dr. Judy, Dr. Berry & Dr. Rubio-Gozalbo

9-9:10 a.m.- Saturday Opening Remarks with Ben Cudzillo

9:10-9:15 a.m.- Galactosemia Angels - we will remember! with Jodie Solari

9:40-10:10 a.m.- Funded Research: Bone Marrow Transplant in a rat model for Classic Galactosemia with Dr. Fridovich-Keil

10:10-10:45 a.m.- Funded Research: Adult Outcomes in Classic Galactosemia with Dr. Berry, Dr. Judy & Dr. Rubio-Gozalbo

10:45-10:55 a.m.- Travel to breakout sessions

11 a.m.-12 p.m.- Breakout Session Series 3

12:15-1:30 p.m.- Galactosemia Foundation Sponsored Lunch

1:30-2:30 p.m.- Breakout Session Series 4

2:30-2:35 p.m.- Travel to breakout sessions

2:35-3:45 p.m.- Coffee & Tea with Breakout Session Series 5 - Roundtable Meet and Greet

3:45-4 p.m.- Welcome to our Final General Session

4-4:10 p.m.- Paul Pruszyński Scholarship Award with Linda Manis

4:10-4:55 p.m.- Panel of Doctors Q&A

4:55-5:10 p.m.- Conference Closing Remarks/2022 Conference location announcement

5-5:30 p.m.- Pick up children from childcare

5:15-6 p.m.- Designated meetup time and space for families to connect

5:15-7:30 p.m.- Dinner on your own

7:30-11 p.m.- Saturday Night Social and Dance

Visit [our website](#) to view any updates to the conference and breakout schedules

Breakout



	Day and Time	Grouper	Damselfish	Fantail	Labrid A	Labrid B	Crystal B
1	Friday 29 1:30-2:30pm	Dr. Gerard Berry What To Expect In 1st 9 Yrs.	Synneva Hagen-Lillevik “Characterization and Composition of Sphingolipids in a Mouse Model of Classic Galactosemia”	Dr. Gomez-Lobo & Dr. Judith Fridovich-Keil Ovarian outcomes and options for fertility preservation in girls and women with Classic Galactosemia	Dr. M Estela Rubio-Gozalbo The galactosemia zebrafish model, what have we learned?	Fundraising 101: how to host a fundraiser and where does it go when donated to G.F.?	Applied Therapeutics Update on ACTION Galactosemia Clinical Program
2	Friday 29 2:45-3:45pm	Anna McGuinness Nutrition management for Galactosemia in Infants and Toddlers	Dr. Nancy Potter- Treating speech and language disorders in galactosemia	Dr. Judith Fridovich-Keil Are long-term outcomes in classic galactosemia progressive?	Dr. M Estela Rubio-Gozalbo GalNet-the Galactosemia Network	Nicole Casale- Financial Literacy, managing one's finances and introduction to credit	Jaguar Updates- Next Steps
3	Saturday 30 11am-12pm	Anna McGuinness Nutrition management for Galactosemia in School Age Children	Dr. M Estela Rubio-Gozalbo -tACS (transcranial alternating current stimulation) as a novel treatment option: a proof of concept study in adult classic galactosemia	Dr. Judith Fridovich-Keil- General update on research from the Fridovich-Keil lab	William Bell- Managing Pharmacy Issues for the Patient with Galactosemia	Debbie Drell NORD - El-Patient Focused Drug Development Meeting Overview (P.F.D.D.)	Dr. Nancy Potter Babble Boot Camp research study - Looking at speech & occupational therapy for babies & toddlers
4	Saturday 30 1:30-2:30pm	Anna McGuinness The older young adult and the changing diet with Q&A	Dr. Gerard Berry Living with Galactosemia as an adult	Babble Boot Camp Team Early intervention for infants and toddlers	Gillian Sapia, RN- Rare Disease Advocacy	Tracey Miller Advocating for Your Child: Educational Planning for Children with Galactosemia	Applied Therapeutics Updates
5	Saturday 30 2:35-3:45pm	No sessions in these rooms, all roundtables and meet & greets in Crystal Ballroom					Networking/Round Tables: Grandparent, Elementary Discussion, Middle School, High School, Transition to adulthood, Meet the Board
Virtual Break Outs		Dr. Ali Parici- Galactosemia and the Spine	Dr. Sandy van Calcar- Bones Basics; Calcium Vitamin D and Beyond and beyond	Dr. Demirbas- Cellular models to study galactosemia	Allstripes- Research and Data Collection		

Visit [our website](#) to view any updates to the
conference and breakout schedules



NEW ALLSTRIPES RESEARCH PROGRAM TO STUDY TYPE 1 GALACTOSEMIA

We partnered with AllStripes, the rare disease research platform, to grow a galactosemia research program with the goal of helping power multiple studies for galactosemia.

We're excited to share the launch of a new AllStripes galactosemia program to increase knowledge about type 1 galactosemia. This research project can help support and inform clinical development of future potential treatments aimed at addressing the genetic root cause of type 1 galactosemia.

Natural history is the usual course of development of a disease or condition in the absence of treatment. This effort will help us understand more about how symptoms present in different age ranges, whether any symptoms correlate with specific lab values and other potential predictors of disease severity.

The de-identified information provided by families in this program may be used to:

- learn more about how type 1 galactosemia progresses in patients over time
- identify and understand important clinical lab values
- support broader type 1 galactosemia clinical development plans for a potential gene therapy

All data shared is de-identified, meaning no personal information like name or phone number is shared.

We are excited to see how this research progresses and look forward to providing you with updates as they become available. Thank you for all you do to move research for galactosemia forward. [Learn more here.](#)

GALACTOSEMIA HANDBOOK AVAILABLE SOON FROM JAGUAR GENE THERAPY

We're excited to share that a new resource will be available soon! In partnership with members of our community and Jaguar Gene Therapy, we have developed a handbook titled, "Navigating Galactosemia Life Stages: A Handbook for the Galactosemia Community." The handbook presents detailed information about how to manage galactosemia, up-to-date diet recommendations, and chapters specifically for each stage of life, including symptoms that may appear and how to be proactive about all aspects of your or your child's care. Recommendations in this handbook from the newborn phase to adult life are based on the international clinical guidelines, peer-reviewed data, and insights from families, medical professionals, and galactosemia experts including Dr. Berry, Dr. Fridovich-Keil, Dr. Longo, Dr. Potter, and others.

Navigating Galactosemia Life Stages

A Handbook for the Galactosemia Community



2022 Edition

This handbook will be available both digitally and in print at the 2022 Galactosemia Foundation Conference, July 28-30. For those who cannot attend, the handbook will be available after the conference on galactosemia.org.

RESEARCH OPPORTUNITY: NATIONAL INSTITUTE OF CHILD HEALTH AND HUMAN DEVELOPMENT (NIH)

Classic Galactosemia can cause premature ovarian insufficiency (POI). NIH is conducting a study to learn why POI happens and if cryopreserving (freezing) ovarian tissue might help to preserve fertility for people with galactosemia.

[Click here](#) to view the flyer on the right.

Has your child been diagnosed with galactosemia?

Classic galactosemia can cause premature ovarian insufficiency (POI)

What is POI: POI occurs when your ovaries stop functioning normally before age 40. In girls and women with galactosemia, POI may occur early in life. When this happens, the ovaries don't produce normal amounts of the hormone estrogen or release eggs regularly. This condition may lead to infertility.

We are conducting a study to learn why POI happens in classic galactosemia, and if cryopreserving (freezing) ovarian tissue might help to preserve fertility in this population

Eligible participants:


- Girls ages 4-12 years old with classic galactosemia who have not yet started puberty.

What will happen during this study:

- Participants will have ovarian tissue removed by a trained surgeon at the NIH. Travel costs will be reimbursed by the NIH.
- Most (80%) of the removed tissue will be cryopreserved (frozen) for the participant's own use later in life. A small piece of removed tissue (20%) will be designated for research.
- The first year of storage costs will be covered by the NIH, but all subsequent storage costs will be the responsibility of the participant's family.

There is no cost for participation and all study related tests and procedures are at no cost to you, beyond the long-term frozen sample storage fees.

Contact:
Veronica Gomez-Lobo, MD at (301) 435-7567, or Email: veronica.gomez-lobo@nih.gov


Eunice Kennedy Shriver
National Institute of
Child Health and
Human Development