

GALACTOSEMIA GAZETTE

“STRONGER TOGETHER” THE 2016 CONFERENCE

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Nicole Casale, Outreach

Kellie Wilcox, Member at Large

We welcome feedback on the Galactosemia Gazette. Please reach us anytime at newsletter@galactosemia.org.



**Galactosemia
Foundation**
Linked for Life.

Dear fellow members of the galactosemia community,
The 2016 Galactosemia Foundation conference from July 14th – 16th in Atlanta, Georgia is just around the corner! If you haven't registered yet, register today. Thank you to those who have already registered. You will be able to register right up until the conference begins – but prices will go up when Regular registration ends on June 15th.

The Galactosemia Foundation Conference Committee and Board of Directors have been working hard to prepare for the conference. We have a great program prepared and I hope that everyone that attends will make connections, learn valuable information, and have some fun.

We have a full agenda of informative general session presentations and interactive breakout sessions planned during the conference. We have increased from five to six breakout sessions for this conference so that you can tailor your conference experience to focus on the topics that interest you wherever you are at in your galactosemia journey. We have also added round table style breakout sessions targeted at different groups from parents of preschool age children through parents and siblings of adults with galactosemia and a session for grandparents /



Scott Shepard, President, Galactosemia Foundation.

extended family of loved ones with galactosemia so that you can network with other people at a similar life stage. On Thursday night, come enjoy some amazing dairy free chocolate and other galactosemia safe candy generously donated by Davide Levine, owner of Illinois Nut and Candy. Illinois Nut and Candy has been a vendor and sponsor at Galactosemia Foundation conferences since the Chicago conference in 2008. You can [pre-order galactosemia safe treats](#) from Illinois Nut and Candy and pick them up at the conference.

Do you have a talent that you would
Continued on page 11...

GF STATEMENT OF ACTIVITY YTD MARCH 2016

Galactosemia Foundation

REVENUE

Child Care Registration	\$	7,557.60	
Conference Registration	\$	18,010.08	
Total G-Groups Registration	\$	9,069.12	
Temporarily Restricted Donations	\$	3,477.00	
Unrestricted Donations	\$	19,258.08	
Total Revenue			\$ 57,371.88

Conference Expenses

Conference Expenses	\$	7,703.99	
Processing Fees	\$	1,070.78	
Dues & Subscriptions	\$	200.00	
Telephone	\$	89.85	
Total Expenditures			\$ 9,064.62
Net Operating Revenue			\$ 48,307.26
Interest Earned	\$	3.99	
Net Revenue			\$ 48,311.25

STATEMENT OF FINANCIAL POSITION MARCH 31, 2016

Galactosemia Foundation Statement of Financial Position As of March 31, 2016

ASSETS

Current Assets

General Fund	\$	159,843.40
EventBright Account Balance	\$	21,033.99
Scholarship Fund	\$	1,185.06
Total Current Assets	\$	182,062.45

Long Term Assets

Research Fund	\$	57,421.26
Total Assets	\$	239,483.71

Research opportunity: Children and Adolescents with Galactosemia

A new study at OHSU is looking at body composition, muscle strength, physical activity and energy needs for those with galactosemia. Body composition studies determine muscle mass and fat mass, which has been found to be abnormal in some individuals with galactosemia. Factors that can affect body composition and energy expenditure include muscle strength and physical activity level. These have not been assessed in those with galactosemia. If you (or your child) have galactosemia and are between the ages of 8 and 20 years, you may be eligible to participate. Study participants will come to OHSU for half a day.

For more information, please contact Sandy Van Calcar, PhD, RD at 503-474-5500 or email vancalca@ohsu.edu

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GG'S AND G-FORCE—ATLANTA HERE WE COME!

By Linda Manis

The plans are set, the trips and entertainment are booked, and the final touches are being made for the GG & G-Force programs at the 2016 Galactosemia Foundation Conference in Atlanta!

Here are a few of the highlights:

- Comedy/Magic Show – Will Brett get chosen as the volunteer again this year?
- Family Feud – Test your knowledge on Galactosemia and “The Survey says ...”
- Yoga – Learn some easy and relaxing yoga moves
- Coca-Cola Museum – Everything you EVER wanted to know about Coca-Cola and more!
- Bowling at the Main Event – Includes Laser Tag, HUGE Arcade, Billiards, and more
- No Whey Southern Café – Back by

popular demand, learn a new recipe and then try it out

- Friday Night – A Grand Night for Talent Together – We hope there will be GGs and G-Forcers on stage too, so we can cheer you on!
- Saturday Night – The famous and fun Galactosemia Foundation Dance

The Conference is only a couple of months away so polish your Southern accents ready for the Peach State (that would be Georgia) and come on down and enjoy some good ‘ole Southern hospitality!

We'll see y'all in Atlanta!



TELL YOUR STORY TO WIN MONEY!

Reprinted from Our Partner TechSoup

At TechSoup, people around the world find the information they need to make smart decisions about technology at their organizations. Join Our Community of Storytellers and Enter to Win up to \$5,000.

Your nonprofit's story matters. Enter TechSoup's 2016 Storymakers contest, where we're awarding \$7,000 in cash prizes and a new camera to the best stories!

If you have a video under five minutes or a five-photo Flickr slideshow, we want to see it.

- Free webinar 1 (recording available)
- Preplanning Your Digital Story from Concept to Story-board
- Free webinar 2 (recording available): Producing a Successful Video Story from Sound to Screen
- Free webinar 3 (recording available): Polishing Your Digital Story to Make it Shine!
- May 31, 2016 — Contest closes
- June 1, 2016 — Judging begins
- June 15, 2016 — Community voting ends

techsoup

About TechSoup

We're a Nonprofit Too! However you do good in the world, there's something for you at TechSoup. TechSoup provides the transformative technology solutions, the digital platforms, and the in-person experiences that enable people to work together toward a more equitable world. It's the generosity of our partners that enables us to help nonprofit, libraries, and foundations like we do. Every contribution helps. We thank all of our partners for their generous contributions.

GALACTOSEMIA'S GOT TALENT



A Grand Night for
TALENT TOGETHER

GALACTOSEMIA FOUNDATION CONFERENCE
Friday, July 15th
7:30 – 9:00 PM
Habersham Ballroom

Share your gifts and talents with your Galactosemia Families!

Do you want to sing, dance, do magic tricks, a sports demonstration, a mime, recite a poem, juggle, play an instrument, tell jokes, do a skit, etc. or find a friend and share your gifts together? The possibilities are ENDLESS!

GUIDELINES:

- Deadline for entry is **June 15th, 2016**
- Open to anyone registered for the conference (all ages) with priority given to those with Galactosemia. Sibling and adults are welcome to participate as well!
- Decide what talent you'd like to share with your Galactosemia families at the conference!
- Each act must be no longer than 4 min. due to the volume of performers
- Only music, lyrics, dress & acts appropriate for family audiences will be permitted.
- Attend a mandatory run-through on Friday night during your assigned time slot between 5:15 - 7:30 pm.

EMAIL: TalentTogether@galactosemia.org with the following pieces of info:

1. Name of Performer (s): indicate if you have Galactosemia
2. Type of Act: (dance routine, musical performance, singing, etc.)
3. Props/Equipment that you'll be bringing: (we will have microphone, stand, piano, sound system, & CD player available)
4. Attach a short video 5 min or less to give us an idea of what you'd like to share

QUESTIONS: Contact Korissa Olson at TalentTogether@galactosemia.org

AN APPEAL FROM THE GENERATION G AND G-FORCE COORDINATORS



Jeannine Quam (L) and Linda Manis (R) at the Coca-Cola Museum in Atlanta.

By Linda Manis

We, Linda Manis (GG) and Jeannine Quam (G-F), have decided with heavy hearts that the 2016 Atlanta Galactosemia Foundation Conference will be our last year coordinating the Generation G (20+) and G-Force (13-19) programs. These programs were started 10 and 8 years ago, re-

spectively as conference programs designed for teens and young adults.

There is no doubt the opportunities we had

to meet and get to know

these amazing young people with Galactosemia have been some of the most rewarding experiences of our lives! This privilege has enriched our lives, the lives of our sons living with Galactosemia and the lives of our families. We, along with our parent volunteers, the GGs, and G-Fs have made many

wonderful friends and lifetime memories, which we will cherish forever! All of you will always be a part of our

"There is no doubt the opportunities we had to meet and get to know these amazing young people with Galactosemia have been some of the most rewarding experiences of our lives!"

"Galactosemia Family."

The GG and GF programs are unique, we know of many similar organizations that do

not have the opportunities that the Galactosemia Foundation offers for teens and young adults. New volunteers are now needed to coordinate and further grow the programs for future conferences. The ideal candidate would observe the groups at the 2016 conference in preparation for leading in future years. We will continue to be available to provide any assistance needed at future conferences.

We encourage anyone who is interested in taking the reins to please contact our Galactosemia Foundation President, Scott Shepard at: scottshepard@galactosemia.org.

SILENT AUCTION AND RAFFLE

By Kellie Wilcox

Announcing the Galactosemia Foundation's Silent Auction and Raffle!!!

The silent auction and raffle will take place all day Friday July 15, 2016 at the Galactosemia Foundation Conference, with bids closing at the "Talent Together"

show in the evening. We are taking donations now of course, but also Thursday and Friday until 6pm during the conference for both auction and raffle. We need many donations to make this event a success so PLEASE don't hesitate. We will have items of different size and value so come

prepared. If you have an item to donate please contact Kellie Wilcox at 903.539.8815 or kelliee@yahoo.com.



ATLANTA ATTRACTIONS

While the Galactosemia Conference, is a one of a kind event, Atlanta has many attractions to offer. Below is some information on many of the top draws in town.

Attractions	Description	Cost	Hours
Centennial Olympic Park (www.centennialpark.com)	The downtown, 21 acre park is a frequent starting point for visitors. The popular Fountain of Rings, is a great place to cool off in the summer.	Free	7 AM – 11 PM
CNN Center (www.cnn.com/studiotour)	The downtown studio is the founding location of the company and is currently home to sister channel HLN. "Inside CNN Tour" visits newsrooms and company history exhibits.	Adults \$15 Seniors \$14 Kids \$12	9 AM – 5 PM
Georgia Aquarium (www.georgiaaquarium.org)	The downtown aquarium features whale sharks, manta arrays, hammerhead sharks, black footed penguins, and thousands of fish.	Adults \$30-\$35 Seniors \$25-\$31 Kids \$24-\$29	10 AM – 5 PM Su-Fr 9 AM – 6 PM Sat
MLK Jr. National Historic Site (www.nps.gov/malu)	Located in Sweet Auburn, just east of downtown, the district is "although and poignant history lesson on the civil rights movement". Sites include Ebenezer Baptist Church, the King Birth Home, the King Center and the tombs of MLK Jr. and Coretta Scott King.	Free	9 AM – 6PM
World of Coca-Cola (www.worldofcoca-cola.com)	Downtown museum dedicated to the soft drink. Includes many historical marketing elements and visitors are given an 8 ounce bottle.	Adults \$16 Seniors \$14 Kids \$12	10 AM-6.30PM Su-Th 9 AM – 6.30PM Fr 9 AM- 7.30 PM Sat
Symphony Hall	Home of the Atlanta symphony orchestra		
Alliance Theater	770 seat live show theatre in midtown	Shows vary	Times vary
High Museum of Art	Midtown's extensive collection includes Degas, Monet, Rodin, Chuck Close, Dorothea Lange, and Georgia O'Keeffe and is considered the best in the southeast.	Adults \$18 Seniors \$15 Kids \$11	10 AM – 5PM Tu-Sat Noon – 5PM Sun
Fox Theatre (www.preserveatlanta.com)	The "Fabulous Fox" in midtown is a lavish 4,000 seat concert hall. Tours Mon, Thurs, and Sat by the Atlanta Preservation Center	Adults \$10 Seniors \$5 Kids \$5 <Cash only>	
Atlanta Botanical Garden (www.atlantabotanicalgarden.org)	This 30 acre attraction in midtown is a "hidden gem" featuring the country's largest permanent orchid collection	Adults \$19 Kids \$13	10AM- 7PM Tu-Sun

“FORE THE CAUSE” TEES OFF FUNDRAISER



Scott Saylor and Jake Saylor teeing off at last year's 7th annual Fore the Cause fundraiser in Richmond, VA.

By Scott Saylor

After attending our first Galactosemia Foundation conference in 2008 and hearing others speak about fund-raising my wife, Kristine, and I decided to take the plunge into fundraising. I had already been involved with many different charities through work so I had some ideas on what we could do. What motivated me in the beginning was hearing the passion for fundraising from others at the conference like the Southard family of Texas. At the time the Southard family had the largest fundraiser for GF and, being very competitive, I wanted to beat them. In 2009 we started “Fore the Cause” a golf charity fundraiser. The first year was small and hectic, yet we were able to raise \$13000. I thought this was an amazing start. We had just moved to Richmond,

Virginia in 2006 and without any family support in the area we were amazed at the giving nature of all those involved. We learned from our mistakes and capitalized on our wins and kept growing our event a little bit each year.

Families involved: It has been amazing to

have other families with children with galactosemia join our event. The Rodgers family (daughter Adrienne) has supported the event by bringing in golfers, sponsors and volunteering. The Stroop family (son Corey) have golfed in our event every year and brought in sponsors. The Berling family has brought in several teams every year. The Wood family has also joined us this year with golfers and sponsors. My Walgreens team has also given a tremendous amount of support with volunteering and raising donation in the community. It has been great to get to know these families and have their support.

Over the years our event has gone on to become a list of events. Some of these have been:

- Selling T-shirts and letting my stores wear blue jeans on certain days throughout the month.

- Selling the local newspaper at a reduced price and the donated \$.05 of each sale to GF.
- Getting my vendors to donate items for raffle or auction.
- My daughter and neighborhood friends having a lemonade stand and donating to GF.

The last three years we have generated more than \$50000 each year to Galactosemia Foundation thanks to the families mentioned above and tremendous support of the community.

To date we have raised over \$250000 for GF. We choose to donate half to general fund and half to research fund because one cannot exist without the other. I also know by donating to GF we



Follow us on Pinterest for updates and information!

are increasing the role GF can make in changing and improving outcomes. I want to thank the entire GF community that holds fundraisers, makes an annual family gift, uses Goodsearch and Amazon Smile, etc. all of these have an impact that continues to strengthen our voice!

MAKING THE MOST OF YOUR FUNDRAISER GOLF TOURNAMENT

By Clayton Aurzada, PGA

As a PGA golf professional for the past eighteen years and parent of a daughter with classic galactosemia, I have had the opportunity to organize and direct thousands of golf tournaments, many of which have been fundraisers. Throughout my career, I have seen some terrific successes and practices. I would like to share some of those with you so that you can put together the most successful event possible. What follows is a list of some of the more creative contest/fundraising ideas I have seen.

On-Course “Fun” Events

Everyone has played golf, so make your event something to be remembered. Longest drive, closest to the pin, longest putt, string, straightest drive, and hole-in-one contests are fun, but they are not that memorable because they are a staple in every golf tournament. Make sure to include these, but also try something different to generate a little more interest.

Fastest Hole- Place a stopwatch on the tee box, have one team member tee off, another hit the second shot, one person chip, one person putt, and stop the clock when the last putt is holed. **Pro tip:** Have volunteers ready with golf carts to transport players to save time.

Throw, Kick, Roll- Have participants buy throw, kick, and roll

tickets, giving them the opportunity to throw, kick, or roll (bowl) their ball without it counting as a stroke. **Pro tip:** This one is hilarious, players remember it, and they come back each year because of it.

The Lifeline- Work with the golf pro to give your contestants the opportunity to “phone a pro” to come out to hit a shot for their team. Players love this, and you can sell more “lifelines” as needed. **Pro tip:** Have the pro “cheat” and hit until satisfied—this ensures that teams get a value for their lifelines.

Triple threat- On a par 3, have teams hit tee shots at the green as usual. On the green, work with the host facility to see if they can cut three different-sized cups in the green (most golf courses can do this: one a little bigger than golf ball diameter, one regulation, and one about the size of a basketball). Cut the small cup in a really difficult position, the regulation cup in a difficult location, and the large one in an easy location. Have teams pay a large fee to go for the large hole, a smaller fee for the regulation, and no charge for the small hole. **Pro tip:** Have the maintenance crew cut the three sizes of cups on or near the tee box so that players can see how small the cups are, enticing them into paying more for the large cup.

Cross Country- Ask the host course if they can create a “new” hole (or even course) for your tournament. Create a hole that spans the widest part of the course, if possible, by teeing off from one tee box and going to a green on the opposite side of the property. **Pro tip:** Try to create a hole where there are multiple ways to get from tee to green for added fun and memories.

Marshmallow Long Drive- On a given hole, have the contestants pay a fee to compete in a long drive contest with a marshmallow while waiting to tee off. **Pro tip:** This one favors shorter hitters, so sell it to inexperienced players.

50/50- On a par 3 (~175 yards), have golfers bet if they can get closest to the hole. The winner for the day splits the pot with your organization. **Pro tip:** Award this in front of the entire field. Many times the winners will donate back their winnings.

Silent auctions- Get as many donations as you can from businesses and individuals. **Pro tip:** Have your volunteers take these out to individual tee boxes to showcase and “sell” them. Many people do not stick around for, dinner, awards, and prizes. Please share any other ideas or contact me with any questions. I would love to help make your event fun and successful: caurzada@yahoo.com.

BEN HUBER RUNS TO RAISE MONEY FOR CONFERENCE ATTENDEES



Ben Huber at Star Wars Half Marathon—the Dark Side—April 17, 2016.

By Linda Manis
 Generation G Ben Huber Raises \$1100 for Paul Pruszynski Memorial Scholarship Fund

After hearing there were Generation Gs and G-Forcers who wanted to attend the conference in Atlanta but could not afford to because of costs, Ben

decided he could help by raising sponsorship money for his Disney Marathon Run. He raised \$1100 to donate to the Paul Pruszynski Memorial Scholarship Fund that provides financial assistance to teens

and young adults with Galactosemia to offset some of the costs for the conference.

Ben started running in 2012 in an effort to improve his mental and physical health. He was challenged by his body's marginal ability to replenish energy stores, but learned how to overcome this prob-

lem by building up stamina and utilizing energy replacements strategically along his route. Ben also learned that running was an effective way to reduce his anxiety and depression as well as increase bone mass ... all of which can be problems for people with Classic Galactosemia.

Ben ran with over 25,000 runners at Walt Disney World and finished the Half Marathon in 2:38. He is looking forward to the Conference in July and seeing all of his friends and meeting new ones!

The Paul Pruszynski Memorial Fund was initiated in 2012, after Paul who, was 23 at the time passed away

suddenly.

If you are teen or Young Adult with Galactosemia in need of financial assistance to offset the conference costs and would like to attend the Galactosemia Foundation Conference in July, please contact:

Linda Manis
 (Lmscript1@aol.com).



Huber with his prize after the race.



Are you signed up for the conference?

It's not too late!

Tickets are still available at
www.galactosemia.org

We hope to see you in Atlanta!

UPDATE: DR. LAI RESEARCH LAB NEWS



Dr. Kent Lai, University of Utah School of Medicine, Pediatric Genetics.

By Dr. Kent Lai

Recent news about the Lai Research Lab at University of Utah School of Medicine

(1) In Nov., 2015, a biotechnology company outside Utah approached the Lai team for possible research collaboration to explore a new treatment modality for Classic Galactosemia.

(2) In Nov., 2015, Professor Kent Lai visited Professor Gerard Berry's Laboratory at Boston Children's Hospital and discussed plans for joint research grant applications in Feb., 2016.

(3) In Jan., 2016, a manuscript

co-authored by the Lai and Berry groups was published in Biochemical and Biophysical Research Communications.

(4) In Feb., 2016, Professor Kent Lai submitted two new grant applications to NIH and Professors Gerard Berry and Estela Rubio were co-Investigators on one of the applications.

(5) In Feb., 2016, additional screening campaigns for galactokinase inhibitors were offered by NIH/NCATS. Such efforts represent an equivalent of US\$500K in FTE and consumables.

(6) In March, 2016, the galactokinase inhibitor (GALK1i) program was selected by NCATS to be included in a program called Research Accelerator, which is aiming to foster collaboration in science among different institutes. It's essentially an online scientific classifieds page started by a group at Yale, and participating institutes (Cambridge, Johns Hopkins, UPenn and University of Toronto, among others) are encouraged to share research ideas

or resources with other groups interested in collaborating or contributing.

(7) In March, 2016, Professor Kent Lai travelled to Bethesda to attend the NIH Small Business Innovation Research (SBIR) Study Section meeting as a member.

(8) In April, 2016, the Lai team presented two posters at the 39th Annual Meeting of the Society for Inherited Metabolic Disorders at Jacksonville, Florida.

(9) In April, 2016, two new biopharma companies have approached the Lai team for discussions on the galactokinase inhibitor (GALK1i) program.



Do you have a success story that you would like to share? If so please e-mail newsletter@galactosemia.org. We are always looking to celebrate achievements and



Follow us on Twitter for updates and information!

HELP WANTED

By GF Board

The Galactosemia Foundation is seeking help from volunteers with specific skills.

Board Members

Galactosemia Foundation will be accepting applications for board positions at the 2016 conference. Please consider joining our team to make a brighter future for all in the galactosemia community.

The board supports a dual mission:

- To educate, support and provide advocacy for those affected by Galactosemia.
- To network with professionals to inspire the treatment and advanced research of Galactosemia

Board duties include supporting families through outreach, empowering fundraising, granting research applications and conducting patient research, overseeing the GF email list, newsletter, webpage and social media accounts, maintaining the GF finances, and organizing the GF conference. The board will be available at the conference to answer questions and speak about open positions. Contact Scott Saylor (scott.saylor@galactosemia.org) for more details.

Natural History Study Project Coordinator

Galactosemia Foundation is working with NORD/FDA to develop an online patient natural history study. "Natural history studies are longitudinal studies that aim to fill research gaps to help medical researchers better understand how diseases progress over time...[They] empower patients and families to help eliminate some of the 'I don't know' in rare disease research, making way for progress." The coordinator will assist with all activities related to the project including: attending NORD/FDA meetings, completing project protocols/paperwork and obtaining approval from the GF board where appropriate, liaising with project medical advisors, testing project IT deployments, etc. Contact Seth Schwartz (seth.schwartz@galactosemia.org) for more details.



CONTINUED...

like to share with the galactosemia community?

On Friday night, come share your gifts and talents and cheer on your fellow members of the galactosemia community at a Grand Night for Talent Together.

On Saturday night we will have our traditional

photo booth, DJ and dance floor. Come down and enjoy the evening with old and newly made friends. Take some crazy pictures to remember the 2016 conference and join the Conga line! We have some special activities planned for our Generation G, G-Force,

and AGERs attendees – including the No-Whey Café, and visits to the Coke Museum and Main Event for bowling, billiards, laser tag, gravity ropes, and an amazing arcade. We also have some great activities planned on-site for our Mighty G

group (ages 9-12) in our childcare.

I am also very excited that there will have several opportunities for attendees to participate in research on-site with Dr. Judy Fridovich-Keil, Dr. Estela Rubio-Gozalbo, and Dr. Sandy Van Calcar.

KEITH TOPPER REPRESENTS GALACTOSEMIA AT NORD CONFERENCE

By Keith Topper

I was invited to participate in a National Rare Disease Day speaking event at Simmons College in Boston, Massachusetts on February 29, 2016. Hosted by the Pre-Health Liaison of Simmons College, the event, which consisted of a panel of four speakers from throughout the rare disease community, was open to students at Simmons College as well as to anyone of the general public. Of the four panelists, three were affected by rare diseases, and one was an activist for rare diseases who works for the National Organi-

“During the event, each panelist was able to share their story and provide information regarding their cause.”

zation for Rare Diseases (NORD). The disorders represented were X-Linked Hypophosphatemia, Frontotemporal Degeneration, and Galactosemia.

After I enrolled into the speaker registry of the National Organi-



Keith Topper (second from left) giving a presentation about living with galactosemia.

zation for Rare Diseases in October 2015, it was not long until a spokeswoman from NORD reached out to me regarding the speaking opportunity at Simmons College. Being that I study at Boston University, I was able to walk over to Simmons College on the day of the event, as it is only a short five-minute walk away from my dorm room.

During the event, each panelist was able to share their story and provide information regarding their cause. Members of the audience were given an opportunity to ask questions in the question-and-answer segment, which concluded the National Rare Disease Day event. The audience, which consisted of about thirty individuals ranging from students to faculty members, was engaging while listen-

ing to the presentations. Once the event concluded, audience members were given the opportunity to have one-on-one conversations with presenters. This ensured that any misunderstandings could have been resolved. In this environment, audience members were able to speak with presenters on a more personal level.

This event would not have been possible had it not been for Andalyn Duke of the Pre-Health Liaison at Simmons College, who organized the event through the National Organization for Rare Diseases. Also, thanks to Simmons College for hosting the event. I was truly honored to be selected to speak on Galactosemia, to be able to raise awareness for Galactosemia (and for all rare diseases), and to be able to tell my story.

GF PARTNERS WITH NORD, FDA FOR NATURAL HISTORY STUDY

**Posted by Jennifer Huron**

Twenty rare disease patient groups have been chosen to develop natural history studies with the assistance of the National Organization for Rare Disorders (NORD) supported in part by a cooperative agreement with the U.S. Food and Drug Administration (FDA).

NORD, the leading, independent nonprofit organization committed to helping people and families affected by rare diseases, today announced the 20 patient groups that will be given the opportunity to develop a natural history study, funded in part by the cooperative agreement, through NORD's Natural History Study research platform.

"Our goal is for the 1 in 10 Americans with rare diseases, most of whom are children, to have a treatment and cure, and we developed NORD's Natural History Study platform to eliminate challenges standing in the way of that target," said NORD President and CEO Peter L. Saltonstall.

"We thank the FDA for its support of NORD and ongoing commitment to rare diseases."

The 20 patient groups are: He

reditary Neuropathy Foundation, Organic Acidemia Association, XLH Network, Inc., CCHS Family Network, Pitt Hopkins Research Foundation, The OMSLife Foundation, Platelet Disorder Support Association, Global Foundation for Peroxisomal Disorders, APS Type 1 Foundation, Scleroderma Research Foundation, Galactosemia Foundation, Desmoid Tumor Research Foundation, International Pemphigus & Pemphigoid, The Morgan Leary Vaughan Fund, Adult Polysaccharide Body Disease (APBD) Research, Bridge the Gap-SYNGAP Education and Research Foundation, United Leukodystrophy Foundation, AMEN-Support (American Multiple Endocrine Neoplasia Support), Lipoprotein(a) Foundation, and Worldwide Syringomyelia & Chiari Task Force.

The groups were determined by a competitive application process reviewed by an internal committee. All diseases represented have diagnostic challenges, limited or no research, and cover a broad range of symptoms and medical specialties, including neurology, cardiovascular, musculoskeletal,

immunology and endocrinology. Selected organizations are all members of NORD.

Natural history studies are longitudinal studies that aim to fill research gaps to help medical researchers better understand how diseases progress over time. They can yield vital information that is essential to clinical trial design, such as biomarkers, demographics, important clinical symptoms, genetic and environmental variables, and patient perspectives.

Rare diseases have posed unique challenges to researchers and drug developers due to small patient populations, lack of data, clinical endpoints that are often unclear, and enrollment and retention challenges for clinical trials. There are nearly 7,000 rare diseases and disorders, 95 percent of which have no treatment. Saltonstall added, "NORD's Natural History Studies project empowers patients and families to help eliminate some of the 'I don't know' in rare disease research, making way for progress."

Ed Note: *The Galactosemia Foundation is very excited to be part of this project with NORD and the FDA. Stay tuned for more information on how you can participate in the study.*

Going to the Galactosemia Foundation Conference in July? PLEASE COME BY OUR STUDY ROOM!

If you are a continuing study participant, please stop by to say hi, meet new members of our team, and make sure we have the most updated contact information for you.

If you have not yet enrolled in our research study please stop by to learn about the study to see if you might want to participate.

If you won't have time to stop by at the conference but want to learn about the study please email Judy at jfridov@emory.edu and we can arrange a time to talk by phone.

Whether you are old or young, male or female, with or without complications - if you have classic galactosemia we need your help!

Participation in our IRB-approved study is easy and free - and NO extra needle stick!



We are conducting an observational research study to define inherited and environmental factors that associate with long-term complications in classic galactosemia. The more people participate, the more powerful the study will be.

What's involved in participation?

After you enroll in the study we will ask you to complete a set of online (or telephone) surveys.

If you have regular clinical blood draws we will ask to collect a small sample of blood for research in conjunction with your next clinical blood draw. There is no cost to you or your insurance company for this procedure and there is no extra needle stick. If you do not have regular clinical blood draws we may ask for a sample of your saliva for DNA studies.

Volunteers who donate blood or saliva may receive free *GALT* genotype analysis, and girls and women who donate blood may also receive free analyses of two hormones related to ovarian function: follicle stimulating hormone (FSH) and anti-Müllerian hormone (AMH).

Questions? Interested?

Please contact Judy Fridovich-Keil at jfridov@emory.edu or 404-727-3924.

THANK YOU! THANK YOU! THANK YOU! THANK YOU!

FUNDRAISING INCENTIVE FOR GALACTOSEMIA FOUNDATION

Galactosemia Foundation would like to invite everyone to participate in the 2016 incentive program aimed at increasing donations that will be used to fund the 2016 Galactosemia Foundation conference. Please note that only donations to the Foundation general fund will be counted toward the incentive program. The general fund covers most of the conference cost. We certainly value all donations, and it is a good idea to split fundraising efforts between the general and research funds.

<p>BRONZE LEVEL - Raise \$750.00 – \$2,499.99*</p> <ul style="list-style-type: none"> • One Conference registration fee waived • Recognized on conference handout • Entered into a drawing for an I-pad mini 	<p>SILVER LEVEL - Raise \$2,500.00 – \$4,999.99*</p> <ul style="list-style-type: none"> • Two conference registration fees waived • Recognized in general session and conference handout • Entered into a drawing for an I-pad mini <p>1st Place** - Reimbursement for up to two nights stay in the conference hotel***</p>
<p>GOLD LEVEL - Raise \$5,000.00 – \$9,999.99*</p> <ul style="list-style-type: none"> • Three conference registration fees waived • Name on conference t-shirt • Recognized in general session and conference handout • Entered into a drawing for an I-pad mini • Reimbursement for up to two nights in the conference hotel*** <p>1st Place** - Reimbursement for one coach round trip airfare ticket***</p>	<p>PLATINUM LEVEL - Raise \$10,000.00 or more*</p> <ul style="list-style-type: none"> • Four conference registration fees waived • Name on conference t-shirt • Recognized in general session and conference handout • Entered into a drawing for an I-pad mini • Reimbursement for up to three night stay in the conference hotel*** • Reimbursement for two coach round trip airfare tickets***

~INCENTIVE PROGRAM DONATIONS ARE ACCEPTED NOW UNTIL JULY 1, 2016~

Donation checks should be made payable to: Galactosemia Foundation
 Donations must be received by **July 1st, 2016**, to qualify for the incentive.

Send to:

Galactosemia Foundation
 P.O. Box 1512
 Deerfield Beach, FL 33443

*To qualify for incentive; you MUST indicate with your donation(s) you wish to have the donation(s) deposited towards the Incentive Program. All donation(s) must be "ear marked" as such and will be allocated to the Galactosemia Foundation General Fund. Money donated to the Galactosemia Foundation Research Fund or Paul Pruszynski Memorial Scholarship Fund do not count towards the Incentive Program.

** *1st Place is defined as the individual that raises the most money within a level. There will be one individual eligible for reimbursement for hotel nights from the Silver Level and one individual eligible for reimbursement for airfare from the Gold Level.*

*** *Reimbursements will be given after a completed expense report has been filled out at completion of the conference. All airfare reservations must be made at least three weeks prior to event in order to ensure lowest possible rate. Airfare portion is valid only from contiguous 48 states.*

Galactosemia Foundation

...LINKED FOR LIFE