Winter 2014/2015

Galactosemia GAZETTE

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Galactosemia Foundation
Linked for Life.

The Galactosemia Foundation is a national, non-profit, volunteer organization whose mission is to provide information, support, and networking opportunities to families affected by galactosemia.

Visit us online at galactosemia.org

2014 Conference was a Social Success

The 2014 international Galactosemia Foundation Conference was a huge success focusing on education, research, fundraising strategies, available resources, new diet awareness, and many other aspects. Socializing with conference attendees, meeting and greeting new and old acquaintances, doctors, educators and families from different countries and states was another opportunity provided.

On Thursday evening, David Levine and Illinois Nuts & Candy again graciously hosted 'A Chocolate Buffet' during and after registration with unique offerings and safe goodies, including a chocolate fountain which many of the children enjoyed for the first time! David and his company has been a generous partner with the Galactosemia Foundation since 2006, and the Galactosemia Foundation is forever grateful for his friendship and generosity. David has generously offered to donate 5% of any sales of corporate holiday gifts to the Foundation if the purchaser uses promo code 'galactosemia'. This opening night social and meet and greet event allowed new friendships to be formed as well as the renewal of old acquaintances from years past.

Also on Thursday, a slideshow of the Dallas 2012 Conference photo booth pictures was looping and various age-appropriate arts and crafts for both children and adults were being diligently crafted with the making of friendship bracelets, sun catchers, painting and coloring books, Play Doh sculptures, etc.; which allowed the kids to intermingle and play together while the parents registered and visited the sales table. It was hart warming to see the older kids helping the younger kids (and adults) with those friendship bracelet looms and glue on do-dads.

After a busy Friday of general and specific learning sessions, the evening social started with a beautiful concert sung by Dale Gillman, a young man from Georgia who happens to have galactosemia. There was also a carnival for both the young and old at heart, as well as a silent auction. All attendees enjoyed the refreshments of snow cones, popcorn and cotton candy, as well as playing putt-putt golf, getting successful splashes on the dunking booth, clown and ring tosses and other carnival game favorites. Plenty of carnival toys, noisemakers, Mardi Gras beads and other goodies delighted all! A special shout out thank you to the Generation G's for their enthusiastic hands-on assistance and for their collection efforts in raising funds for the Paul Pruszynski Memorial Conference Scholarship Fund.

After even busier Saturday sessions, a DJ and dance closed the conference with leis, silly hats, sunglasses, glow in the dark bracelets, very active hula hoop and limbo contests, and plenty of sweet goodies for refreshments! The photo booth was very popular and 615 pictures were taken with duplicate memory souvenir photos given out. Also during the Saturday party, the Galactosemia Foundation honored Michelle Fowler's retirement as President and for her untiring fourteen years of volunteer service to our Foundation.

The Conference Committee wishes to thank all its board members, doctors, educators, volunteers and attendees and hopes you will join us in Atlanta on July 14-16 2016 for another successful Galactosemia Foundation Conference!

The Galactosemia Foundation is a non-profit charitable organization. Founded in February 1985 by a small group of mothers in New York, We realize the need for further information and networking between affected families and professionals. Metabolic Clinics across the nation continue to assist The Galactosemia Foundation in researching families and information. Today our mailing list includes over 1000 families and extended families, professionals and clinics, media groups, donors, and numerous international contacts. Objectives and functions are achieved by unpaid volunteers.

TThe Galactosemia Foundation publishes two newsletters per year. These newsletters vary depending on the conference years! If you have an article or a fundraising event that you would like to have published in the newsletter please submit it to: **newsletter@galactosemia.org**

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

A Letter From the President

Dear Galactosemia Family,

I can't believe it's already been a few months since taking the leadership role and transitioning into the duties of president of the Galactosemia Foundation. Like many things in life, a new role is both exciting and frightening at the same time.

My family and I are thankful for the dedication and perseverance of past leaders and volunteers, who created, sustained, and grew the Galactosemia Foundation from its beginings as Parents of Galactosemic Children (PGC).

Whether you are a new member of our galactosemia family, seasoned parents of galactosemic adults, or somewhere in between, this is your foundation. The Galactosemia Foundation is a 100% volunteer organization funded entirely by donations and fundraisers. Thank you to all of you who are active volunteers, fund raisers, and donors. As you will see from the treasury report in this newsletter, the 2014 conference cost the organization over $250,000. The attendees paid about $64,000 of this cost in registration fees. The $192,000 difference – about $360 per attendee – was paid for by your donations and fundraisers. As 2014 draws to a close, please consider making a tax deductible donation to the Galactosemia Foundation and begin planning your next fundraiser!

February 2015 will mark the 30th anniversary of the founding of our organization as Parents of Galactosemic Children (PGC). As we enter our fourth decade, the board and I are dedicated to:

1. Continue our mission to "educate, support and provide advocacy for those affected by galactosemia" and to "network with professionals to inspire the treatment and advanced research of galactosemia".
2. Make changes to improve the financial stability of the Foundation (such as going to electronic newsletters and likely raising the 2016 conference registration fee).
3. Increase the frequency of our communications by filling the vacancies and adding to the members of the Communications Team

- Scott Shepard

New Challenges and Improvements

I'd like to share with you recent news, challenges and improvements we will be working on as a team:

- Michelle Fowler announced that she was retiring as President of the Galactosemia Foundation at the 2014 conference in Orlando. Michelle served on the Board of directors for over ten years. Thanks to Michelle's leadership, we are no longer just parents of children with galactosemia, but also children, teens and adults with galactosemia, grandparents, husbands, wives, brothers, sisters, friends, and children of people with galactosemia.
- Diane Flynn-Brake has also retired after six years on the board, serving most recently as Secretary.
- Paul Fowler has announced his plans to retire from the Board at the end of 2014 after serving more than eight years as Treasurer. Paul has agreed to remain as Treasurer through the end of 2014 to ensure a smooth transition to the new treasurer.
- Additionally, Angie Canul and Karen Haley, who have been instrumental in editing and creating the professional look and feel of our newsletters since 2008 are stepping down from the Newsletter team after publishing this edition.
- Since the conference, our focus has been on filling the vacancies on the board by these retirements and key leadership roles on the committees. I want to share the process that we went through to select our four new board members. Scott Saylor led the effort to fill open board positions from among candidates that submitted volunteer interest forms at the 2014 conference. We had 37 volunteer interest forms submitted during and after the conference. We had many strong candidates with impressive qualifications – thank you to everyone that expressed their interest in becoming more involved in the foundation. Among those submitting interest forms, there were 14 people that were interested in and completed applications to be on the Board of Directors. The Board reviewed all of the applications and narrowed the pool to five candidates that were then screened with phone interviews. We then offered Board positions to four of the candidates.

Please join me in offering a warm welcome to our new board members: Nicole Casale, Seth Schwartz, Lisa Spiro, and Kellie Wilcox. Lisa joins the board as the Treasurer Elect and is working closely with Paul Fowler to transition into the role of Treasurer in 2015. The other new board members have all joined the board as Members at Large.

Also, please join me in congratulating Scott Saylor and Andrea Topper on being elected as the new Vice President and Secretary respectively. I look forward to working closely with the new officers and Board members in 2015 and beyond.

Help Wanted

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

Editor – member of the Communications Team. The editor will plan the contents of Galactosemia Foundation publications. The editor will also prepare, rewrite and edit copy to improve readability, and correct errors in spelling, punctuation, and syntax. Work with the Graphic Designer to allocate print space for story text, photos, and illustrations according to space parameters and copy significance, using knowledge of layout principles.

Graphic Designer – member of the Communications Team. The graphic designer will create page layouts for our newsletter and other Galactosemia Foundation publications. The graphic designer will determine the size and arrangement of copy and illustrative material and select the style and size of type for printed materials. Working with the editor, the graphic designer will assemble final layouts for printed materials. Our newsletter is currently laid out using QuarkXpress. The ideal candidate would be an expert in QuarkXpress or comparable desktop publishing software and own their own copy of such software.

Web Developer – member of the Communications Team. The web developer will design and develop the Galactosemia Foundation conference registration website for the 2016 conference.

System Administrator – member of the Communications Team. The system administrator will manage the Galactosemia Foundation's Microsoft Office365 Technology grant – including Office 365, Exchange, Lync, and Sharepoint.

Writer/Reporter – member of the Communications Team. The writer/reporter will interview people, build contacts and sources for future stories, fact check information given by sources and create copy for Galactosemia Foundation publications.

Galactosemia Foundation's 2014 "Teaming Together For A Bright Future" Orlando Conference Treasury Report

INCOME:

Adult Registration		$38,515.77
Child Registration		$10,287.94
G-Force Registration		$6,590.08
Generation-G Registration		$6,872.95
AGERs Registration		$1,550.74
Total Conference Registration Income		$63,817.48
Other Income:		
Conference Fundraising Incentive		$101,824.86
Conference Donations		$12,119.33
Conference Sales & Raffle		$14,408.42
		$128,352.61
Total Income:		**$192,170.09**

EXPENSES:

Food and Beverage:	Breakfasts	$35,464.70
	Lunches	$68,896.76
	Breaks	$16,161.80
	Registration and Socials	$11,659.23
	Childcare Snacks & Drinks	$4,885.36
Travel & Expenses:	Lodging (Speakers, Childcare, Board)	$17,376.45
	Travel – Speakers	$5,623.37
	Travel – Board	$6,171.26
Audio/Visual Contract		$25,888.03
Childcare Contract		$17,600.00
Pre-Conference Planning Meetings		$14,705.68
Generation "G", G-Force & AGERs Programs		$10,553.28
Office Supplies, Printing & Postage		$4,232.57
Registration Materials, Signs & Banners		$4,261.90
Fundraising Incentive Program		$3,931.88
Sales Items		$3,162.61
Entertainment:	Friday Night Social	$1,507.68
	Saturday Night Social	$1,595.00
Insurance		$398.00
Total Expenses:		**$254,075.56**

The $63,817.48 conference registration fees paid by attendees only covered about 25% of the total cost of the conference. The remaining 75% of the conference expenses ($190,258.08) was covered in part by $128,352.61 in other conference income shown above. The rest of the conference expenses ($61,905.47) were paid for out of the General Fund. After all conference expenses were paid, the General Fund balance was $11,908.37. We need your donations and support to build our funds back up for the 2016 conference.

Account balance as of 11/16/14

- General Account $18,615.09
- Paul Pruszynski Memorial Scholarship Fund $839.79
- Research Account $94,356.65

Our 2016 conference incentive program is now open! This is a great way to raise funds for the 2016 Atlanta conference and can also help you offset some of your travel expenses. Please keep in mind that in order for your donations/fundraising to be considered for the incentive program they must be marked and deposited into the General account. Full details will be published on the website. If you have any questions on the incentive program please email me. In order for GF to continue to host conferences donators need to remember the General account. Help us reduce costs by registering with our on-line database which will allow families to update addresses, phone numbers, and email. If you would like a complete breakdown of the conference report please send a written request to me at paul.fowler@galactosemia.org.

Paul Fowler
Treasurer

Treasurer Elect - Lisa Spiro

Hi Everyone,

My name is Lisa Spiro and I am excited to serve the Galactosemia Foundation as Treasurer Elect. I have over 20 years accounting and finance experience and currently work as a controller for a healthcare company that manages mental health hospitals. I have my Masters in Accounting and have passed the CPA exam. I have also served as treasurer for various school organizations over the years. I will be assuming the responsibility of the treasurer from Paul Fowler over the next few months. I look forwarding to working with the board to further the goals of the organization.

I live in South Florida with my husband, Allan, and my three daughters, two who have classic galactosemia. I have been a part of the galactosemia family since 1995, when my second daughter, Megan was diagnosed at 6 days old. My first conference was in Ohio in 1997 and I was thrilled to finally meet other parents who were experiencing the same issues and facing the same fears of the unknown. When I was pregnant with my third daughter, Amanda, the results of the amnio indicated that she also had classic galactosemia. Although I did not want either of my girls to have galactosemia, I was comforted by the thought of them having each other.

Megan and Amanda are now a senior and a junior in high school, respectively and my oldest daughter, Kayla, who is a carrier, is now a senior at the University of Central Florida. It's amazing how quickly the years fly by. I couldn't be more proud of all my girls and the individual challenges they overcome by having galactosemia or being a sibling.

Through the years I have attended 7 conferences and have enjoyed not only all of the great information provided by the medical professionals but also meeting other parents and hearing the stories. I believe that the Galactosemia Foundation plays a critical role in helping parents and children; initially by hosting the conferences, and then by awarding the research grants. I am honored to be allowed to a part of the team and eager to give back while continuing to advance the mission of the organization.

Welcome New Board Members

Greetings from Kellie Wilcox

Our story starts in 2002. Our first son, Landon, was born with galactosemia, dwarfism, a hole in his heart, no pituitary, adrenal, or thyroid glands and the list goes on. My husband, Jason, and I know now that Landon was brought into and taken out of our lives for many reasons, one of which was to alert us to galactosemia. For two years I researched and studied galactosemia. I had lost my son and if I was ever going to have another child, I was going to know everything I could know the next time! Boy did I learn a lot about genetics! In 2004, a perfect little baby boy named Paden, was born! We opted to not do an amino, started him on soy at birth and waited for the test! Positive CG results really didn't surprise us and we were ready. Paden was an excellent baby, no speech or motor issues as an infant/toddler, no learning difficulties or cognitive delays at school age. Paden is now 10 years old and entered the 5th grade and while he is extremely social and a well-rounded class clown, he is falling behind. Everything in school is a struggle!

We waited 6 long years to decide if we even wanted to try for another child with the difficulties we had already experienced. In May 2010, our daughter Keelee, was born and thankfully she is a carrier! June 2010, when Keelee was seven weeks old, my sister was killed in an auto accident and in Sept 2010 her husband was also killed in an auto accident, leaving a 6 year old daughter (five months younger then Paden) and a 12 month old son (eight months older then Keelee). Within a four month period our family went from three to six! That was four years ago and we wouldn't have it any other way!

My husband taught for eight years as an agriculture teacher for Rusk ISD and now is on his seventh year as the Assistant Principal at the high school. I am on my 16th year at a small credit union in our town, eight years as the CEO. We are both from small towns in East Texas and currently live about fifteen minutes from where we work, in New Summerfield. We live on several acres and raise different breeds of livestock. We participate as a family in 4-H and softball leagues year around. Our kids are our life! I am sure all of you know exactly what I mean.

I am extremely excited about the opportunity to serve on the Galactosemia Foundation board. I hope to bring much support and new perspective to the board, as I also hope to learn a great deal from the foundation. Classic Galactosemia effects our life every day, through our son, and I want to help him and others like him, however I can!

Nicole Casale

Hello everyone in the galactosemic community,

My name is Nicole Casale and I am from Latham, NY. I am so excited to be a member of the board and am looking forward to contributing in any way that I can.

My husband Nick and I have two boys Noah, age 3, and Joseph, age 2. Joseph was born in March of 2012 and diagnosed at day five of life with galactosemia. Today he is doing great, but like so many others had a rough start, contracting meningitis, which led to profound hearing loss in his left ear. Noah does not have galactosemia, and we do not know if he is a carrier or not.

For work, I teach financial education to adults at Troy Rehabilitation and Improvement Program, a non-profit organization. In my spare time I enjoy running, cooking, and fundraising. I have a few fundraisers under my belt and look forward to planning more in the future.

Seth Schwartz

Hi members of the Galactosemia Foundation,

I'm Seth Schwartz from Toronto, Canada and I am thrilled to have been invited to join the board of the foundation. I look forward to serving, you, the members, in fulfilling the foundation's mandate of raising galactosemia awareness, providing education, and supporting research on the condition.

My wife Charlotte and I have two sons, Isaiah and Rivers. Isaiah was diagnosed with classic galactosemia in 2011 at 10 days of life. He is currently receiving occupational therapy, but is otherwise in good health. Rivers does not have galactosemia.

Professionally, I am the Manager of Pharmacy Business Analytics for Shoppers Drug Mart, the largest retail pharmacy in Canada. I am also the VP on the board of directors for the Creative Preschool of East Toronto, which Isaiah attends, a role that has given me some experience in providing leadership to a non-profit. In my spare time I am a huge basketball fan and a fitness enthusiast.

Outreach Team Spring 2014 Update:
Robyn Meek

Hello galactosemia family, it's time for another newsletter! This gives us a great chance to remind you of the work we do, and introduce ourselves to those unaware of what the Outreach Team is all about!

The Outreach Team is organized by a few passionate parents whose goal is to welcome new families into our galactosemic community. What exactly do we do? Here are some of the things we have done this past year:

- We answer LOTS of emails, phone calls and text messages from new families searching for help.
- We manage a Facebook discussion board geared towards new families and their concerns, in the past few months this group has grown from under 150 members to over 300. (Find us at: "Galactosemia Outreach Team Discussion Group") *We have a pinned post specifically for introducing your family and location, find new contacts in your area by browsing through your fellow families that have posted there!
- We remain closely connected with newborn screening advocates and the Save Babies Through Screening Foundation.
- We provide guidance and advice to new families struggling with a new diagnosis.
- We have helped new families find local contacts for support and friendship.
- We have helped new families learn about their local support services for early intervention.
- We have helped new families find geneticists and dietitians in their area.
- We have helped decipher ingredient lists and shared dining out strategies.
- We have talked with new families from India to Indiana just in the past year.

If outreach work has taught us anything, it is that we are NOT ALONE at all. Every week, new families are finding us from across the globe. The ways that we can help each other are endless. We have some amazing go-to contacts scattered across the country that we rely on heavily when a new family arrives. If you would like to be involved in welcoming new families, sharing your experiences, and sending out a warm hug (be it in person or virtually), please contact one of us! Creating connections is our greatest asset!

Here are the local US support groups that the Outreach Team is aware of (if you have one that is not listed, please let us know):

- GAMA (Galactosemia Association of Midwest America) www.galactosemiamidwest.com
- GFMN (Galactosemia Families of Minnesota) http://galactosemia.webs.com
- GFSS (Galactosemia Families of the Southern States) Contact: Jo Beth Southard, jobethsouthard@yahoo.com

Future Outreach Goals:
On a more personal note, in 2009, our family was introduced to galactosemia with our first baby girl. I remember we frantically scribbled down g-a-l-a-c-t-o-s-e-m-i-a as we received that initial newborn screening call, the word and the disorder were so foreign to us. I remember receiving our first galactosemia newsletter when our daughter was just a few months old and I was SO overwhelmed. Our family was fortunate to have a wonderful local support group that scooped us up and really took care of us that first year. Our family is driven by and forever grateful for two things:

1. Timely newborn screening results and an attentive pediatrician.
2. Our local galactosemia support families.

I truly believe a large focus of our outreach efforts need to be directed towards the newborn screening process and the education of hospital staff. With these things in place, our galactosemic babies have the best start in life possible.

Five years after our initial diagnosis, I am still receiving emails from new families who are not getting their results for two weeks, two months, or worse. In 2009 we had a suspected galactosemia diagnosis by day three and had been advised to switch to soy formula. Timely results and action are possible, but unfortunately, not a reality across the globe. I am still hearing from families who were told "galactosemia is so rare, it's surely a false positive, continue breastfeeding". These stories are so unacceptable and my goal is to see every baby have the best chance possible. If you have any suggestions to improve these processes either locally or nationally and would like to help us find the best way to improve the turnaround time on galactosemia diagnosis and care – please contact us and share your ideas.

The entire outreach team - outreach@galactosemia.org
or individually:
Robyn & Jeremy Meek – Illinois, meekfamily1@gmail.com
Tara Tanella – New York, ttanella@gmail.com
Tara McCoy – Kentucky, Tara.L.McCoy@ehi.com

CONQUERING GALACTOSEMIA THROUGH STEM CELL RESEARCH

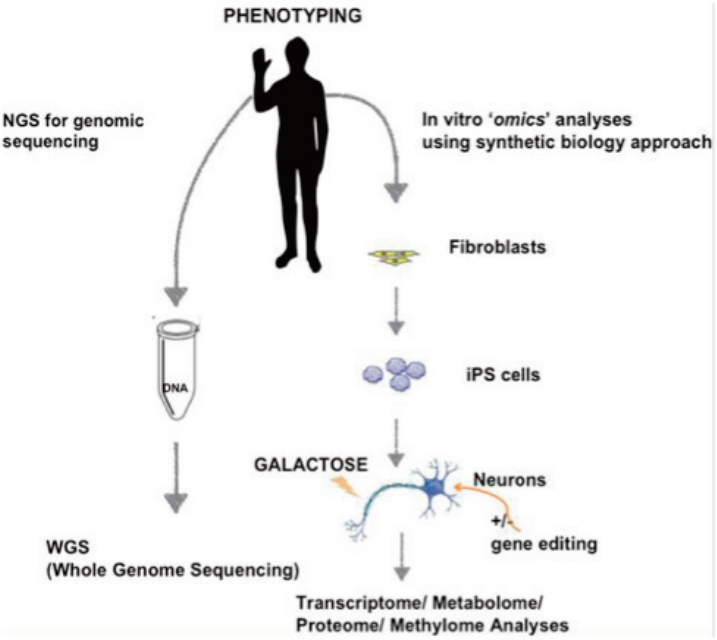
Gerard T. Berry, Didem Demirbas, Cynthia S. Gubbels, and Xiaoping Huang

Boston Children's Hospital has created a special environment for the care of patients with galactosemia and research on this rare disaease. This includes the Center for Galactosemia and the Manton Centor for Orphan Disease Research. In this update, we would like to focus on the research that is ongoing at Boston Children's Hospital/Harvard Medical School. In the Berry laboratory, we are maximizing the opportunities that opened up with the successful creation of galactosemic induced pluripotent stem (iPS) cells.

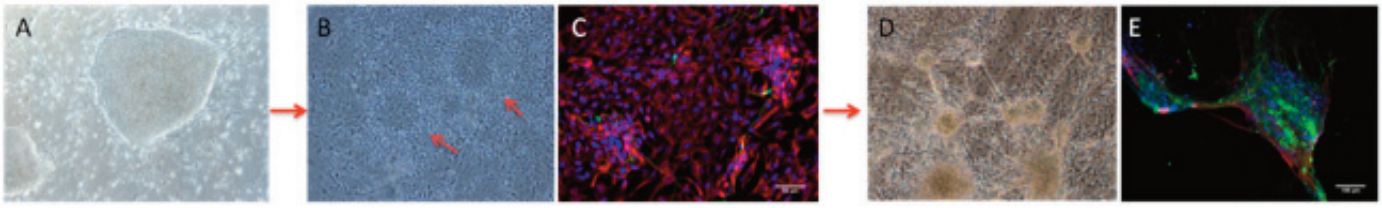
We collaborated with Boston Children's world-renowned stem cell expert George Daley, MD, PhD and were able to successfully generate different lines of induced pluripotent stem cells that carry the Q188R/Q188R and Q188R/Q188P genotypes. Please note that Q188R mutation is the most common classic galactosemia mutation in Caucasians. Q188P is a recently discovered new mutation that behaves like Q188R. Furthermore, we were able to turn these stem cells into neurons. This gives us a unique chance to study galactosemia in the disease relevant cells.

We have exposed the galactosemic iPS cells to calibrated doses of galactose. These carefully calibrated set of experiments allowed us to quantify the galactose metabolites in these cells using our state-of-the-art LC-MS/MS techniques. The LC-MS/MS instrument is a mass spectrometer and allows investigators to perform state-of-the-art analyses for the molecular composition of various chemicals. In our studies, galactosemic iPS cells have accumulated higher levels of galactose and galactose-1-phosphate (the substrate of the GALT reaction) compared to the control cells but on the contrary UDPgalactose (the product of the GALT reaction) remained flat in galactosemic cells. These experiments demonstrate that the biochemical galactosemia phenotype is reflected in these iPS cells that lack GALT enzyme activity. We are in the process of expanding our analyses on the iPS cells, which -we believe- will give us clues on the effect of GALT deficiency at the prenatal period.

We are also implementing the latest cutting edge research techniques in our experimental approach to understand galactosemia pathophysiology. We are using the recently developed CRISPR/Cas9 gene editing tool for introducing the GALT mutations in the control background as well as correcting the Q188R mutation back to wildtype. This experimental study will help us generate disease-free iPS cells that are otherwise identical to our patient cell lines and they will serve as control lines in our experiments. This will reduce the 'background noise' that occurs due to genetic differences between individuals in all of our analyses.



As we broaden our knowledge on these galactosemic induced pluripotent stem cells, we are learning more about the basic mechanism behind the effect of galactose in the absence of GALT. In addition to this, the iPS cell approach in conjunction with whole genome sequencing allows us to ask another scientific question: why the complications of galactosemia are different between different individuals? We hypothesize that the specific GALT gene alteration, together with the behavior of modifier genes and epigenetic effects all contribute to the types and severity of complications that patients experience. The support from Galactosemia Foundation enabled us to test this hypothesis by creating stem cells from individuals with variable phenotypic outcomes and comparing the findings with the whole genome sequencing data. We have already completed sequencing of one adult patient with galactosemia. We are excited to tackle these questions with the cutting edge research methods and we believe that any advancement in our understanding of the effects of GALT deficiency in fundamental cellular functions will bring novel insights that guide treatment strategies and development of new therapies. We are grateful for the support and encouragement from Galactosemia Foundation.



Differentiation of galactosemic iPS cells (A) into neural progenitor cells (B-C) and early stage neurons (D-E)

October 2014

Dear Galactosemia Foundation Families:

First, a huge **THANK YOU** to the organizers and all the wonderful families who made the 2014 Galactosemia Foundation Conference in Florida the amazing experience that it was. What a treat to reconnect with so many old friends and meet so many new, and a special thank you to everyone who came by our study room to sign up – or just to say hi! It was our privilege to be there with you, thank you!

Many volunteers and families like to keep up with current research findings, so I will take this opportunity to give you a list of our relevant publications from 2014. If you would like to receive a copy of any of these articles please just let me know (jfridov@emory.edu or 404-727-3924).

Of course, other studies are currently underway or just getting started... never a dull moment! As always, the more people who participate in our research, the more meaningful the results will be. I will put a miniature version of our current recruiting flyer here – please feel free to pass this along to anyone you think might be interested. Thank you.

- Jumbo-Lucioni, PP, EL Ryan, ML Hopson, HM Bishop, T Weitner, A Tovmasyan, I Spasojevic, I Batinic-Haberle, Y Liang, DP Jones, and JL Fridovich-Keil (2014). Manganese-based superoxide dismutase mimics modify both acute and long-term outcome severity in a *Drosophila melanogaster* model of classic galactosemia. Antioxidants and Redox Signaling Forum 20(15):2361-71
- Pyhtila, B, KA Shaw, S Neumann, and JL Fridovich-Keil (2014). Fifty years of newborn screening for galactosemia in the United States: Looking back, looking around, and looking ahead. J Inherit Metab Dis Reports, 2014 Apr 10. [Epub ahead of print]
- Li, Y, X Huang, L Harmonay, Y Liu, M Kellogg, JL Fridovich-Keil, and GT Berry (2014). Liquid Chromatography Tandem Mass Spectrometry Enzyme Assay for UDP-Galactose 4'-Epimerase: Use of Fragment Intensity Ratio in Differentiation of Structural Isomers. Clinical Chemistry 60(5):783-90
- Tran, T-TV*, Y Liu*, M Zwick, D Ramachandran, D Cutler, X Huang, G Berry, and JL Fridovich-Keil (2014). A *de novo* mutation in galactose-1-P uridylyltransferase (GALT) leading to classic galactosemia. J Inherit Metab Dis Reports, accepted.
- Lynch, ME, NL Potter, C Coles, and JL Fridovich-Keil (2014). Developmental outcomes of school-age children with Duarte galactosemia: A pilot study. J Inherit Metab Dis Reports, accepted.

Best wishes,

Judith L. Fridovich-Keil, PhD
Professor, Department of Human Genetics
Emory University School of Medicine
EMAIL: jfridov@emory.edu,
TEL: 404-727-3924

CALLING ALL PEOPLE WITH CLASSIC GALACTOSEMIA
...we need the whole gang!

Did you ever wonder why many, but not all, people with classic galactosemia experience long-term problems even if they were identified as newborns and have always excluded galactose from their diet? Do you wonder if something might be done to prevent or reverse these problems? We are working to find the answers – and we need your help!

If you have classic galactosemia, please consider volunteering for our research study. Whether you are old or young, male or female, with or without long-term complications – we need your help!

Participating in this important study is easy, free, and requires no extra needle stick.

About the study:

- This study "Bases of Pathophysiology and Modifiers of Outcome in Galactosemia" has been approved by the Emory University IRB (Emory eIRB00024933, Principle Investigator: JL Fridovich-Keil)
- This study is modular – you can participate in only those parts you want to.
- No extra needle stick or invasive procedures required.
- You can participate long-distance – no travel required.

Questions? Interested?

- Please contact Dr. Judy Fridovich-Keil, the principal investigator of this study, at jfridov@emory.edu or 404-727-3924.

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

Need for Discussion/Networking with Parents of Adults with Classical Galactosemia

I would like for parents with adults who have classical galactosemia to visit between now and our next conference (2016) to discuss: (1) how their young adults with galactoesmia are doing and why we don't hear much from those adults who are doing well?; (2) what education level have the adults achieved? (3) are the adults with galactosemia working? (4) of those who are working, how many adults with galactosemia are plumbers, electricians, biologists, ...etc.? (5) have the parents of adults with galactosemia thought about what happens to your adult children when you pass away?

Regarding the latter, I was visiting with a mother of an adult with Classical Galactosemia at our last meeting. We discussed what would happen to our children (now adults) after we pass away. While I write this, I believe my son is fully capable of taking care of himself now and in the future. However, I would rest easier if there was some place where adults with galactosemia who are not totally independent could live, work, and interact with others in their home and in the community. There are some places in a number of states that have specialized in adults with special needs.

If you would like to have a running dialogue on adults with galactosemia (to include one through five above), and you think it is important for parents of adults with galactosemia to meet at the 2016 conference in a separate session with agendas/forums/round-table-discussions, please contact us by email.

Nate and Rhonda Bauer
nate.bauer@suddenlink.net

2014 SILENT AUCTION AND RAFFLE

Kellie Wilcox

Thank you everyone for the wonderful second biannual Galactosemia Foundation silent auction and raffle! We had items donated by galactosemic support groups, companies, and individuals. It would not have been possible without all of these items and the volunteers that worked so hard to get them donated. We had 61 auction items and 23 raffle items. There were over 60 people signed up to participate in the fun on Friday night. The silent auction and raffle profited over $6,000! That is a $2,000 increase from the last conference! That would not have been possible without each of you. All of those funds helped to cover the costs of the 2014 Galactosemia Foundation Conference! I hope you all had as much fun as we did with the silent auction. If anyone has any comments or suggestions for the next silent auction, don't hesitate to email me.

kelliekee@yahoo.com

Message from Scott Saylor

I want to continue my message from the 2014 conference. I truly know we are stronger together as one foundation. When my family started fundraising we wanted to make the biggest impact possible. We knew by donating to both the research fund and the general fund we would make GF stronger and increase its abilities to make an impact on the future. GF has so much to accomplish and getting support from our families is important to help us accomplish those goals. I think everyone needs to realize when we stand together as one voice more people will take notice. Researchers, drug companies and companies that could support our cause will take notice. As they do take notice this will advance research and increase educational opportunities which are the mission. As an individual I know much of this research will not impact my child. However many years from now as I look back on my life I want to be able to say that I had an impact on galactosemia.

Your impact no matter how big or small is changing the Galactosemia Foundation for the better. Please consider starting your own fundraiser, starting an annual family gift or asking companies to donate to GF. Together we can change the future.

Please Remember...

Stories in this newsletter are personal experiences and opinions and are not necessarily the opinions of The Galactosemia Foundation and/or medical professionals. They are simply offered to share information & ideas. Always check with your genetic professional and dietician before making any changes to your child's diet or healthcare.

THOUGHTS BEFORE FUNDRAISING

Charlotte Schwartz

Isaiah Schwartz was born with classic galactosemia in March 2011. His mother, Charlotte Schwartz attended the 2014 Galactosemia Foundation conference in Orlando, where she gained a much deeper understanding of what the diagnosis means. Upon returning home, she challenged herself to help advance the galactosemia community and decided on fundraising towards a lofty goal. Below are her thoughts the night before her fundraiser concluded:

Here goes nothin'...

Every time I sit down to write this, tears start to flow and I can't get through to the end; my hands shake and my tears strike the keyboard below. The result a blurry, incoherent "thank you" of sorts.

But it's time to rest ahead of tomorrow. And so here is my raw, unedited and all-encompassing thank you, before it's too late.

I have been very active for the greater part of the past 10 years - part of my daily routine - like eating or breathing. What initially began as a plan to get into shape and to be healthier overall has certainly evolved with the passage of time.

But running has always been there. And after Isaiah was born, it became the most affordable kind of therapy there was. After Isaiah's diagnosis, my head felt like it weighed a ton; my body leaden and exhausted much of the time, weighed down by the questions. Weighed down by unprocessed guilt, grief, and the gravity of our new "normal".

I needed a way to reconcile that, so when he was six weeks old and I got the "green light" to exercise again, I laced up my Nikes and I went for a run.

The brisk April air was intrusively refreshing. With every strike of pavement, a piece of my frustration fell away; a part of my misplaced anger dissipated.

With each natural swing of my arms, I propelled my body forward. There was no greater metaphor than that, in that time. Moving forward, not looking back. Not worrying about what I may have missed.

Fast forward three and a half years - In July, I posted a link to the fundraising page I set up [with the local children's hospital's website]. I set a lofty goal of $10,000 and laughed to myself when I typed all of those zeros into the form on the website where it asked me to enter my "Goal". And then I put it out there - in the Internet-Universe - and I walked away.

A few hours and a few pings from my iPhone later, I had two donations from two amazing people - Carol Babcock and Claire Washburn. And with that, I was at 1% of my target. $100 raised. Grateful for their donations, I went on to the rest of my days' work and giggled to myself about all those zeros again later on.

Over the last 18 weeks, I've trained hard, thought hard, tossed aside much of my own negativity and silenced a lot of the voices (figurative and literal) that told me this couldn't be done.

Because $15,130 - that's why. Not a small feat by any stretch- but for galactosemia- your support is epic. It mimics the resultant cannonball splashes of 15,130 people jumping into a pool built for 10.

And I can't thank you enough for that, and I can't explain to you what it really means - not yet. You took the time to click the link, to read the story, and then you gave generously and without reservation. You gave part of the money that you work so hard for, to this disease that you've never even heard of. I feel so much because of that, but I can't quite articulate it yet.

I would say that I feel gratitude, but for the fact that gratitude is what you feel when you receive a compliment or a great birthday gift. I feel like I've been given 15,130 great birthday gifts, and it's not even my birthday.

I would say that I feel lucky, but lucky is how you feel when you find $10 in your winter jacket from last year. I feel like I've found 15,130 winter jackets with $10 in every pocket.

But because that wasn't enough, [the hospital] decided they would establish the first-ever Galactosemia Research Fund - dedicated dollars for this rare disease. An incredible outcome that exceeds my wildest and most obscene expectations.

One of my life's greatest pleasures thus far has been watching that little red thermometer on my fundraising page creep upwards until it eventually burst under the pressure of your generosity, shattering my expectations ten thousand times over.

But now tomorrow is nigh. It's time to rest my head for tomorrow's endeavour. I'll bid you goodnight and thank you, from the bottom of what was, for a time, a heart a bit sunken and a bit shrunken. I thank you for all you have made possible. I thank you for joining me in making an actual piece of history here on our little piece of this rock.

And finally, to my sweet, amazing boy Isaiah - this next part is for you. One day I hope to tell you a story about how mommy ran and ran and ran until, one day, there was a cure for this disease called Galactosemia.

I love you fiercely, and I'll see you at the finish line, my love.

Another Memorable Time for Generation Gs and G-Forcers

Linda Manis

We had the largest group ever of Generation Gs and G-Forcers from all over the country; let's make that from all over the world, two GGs girls came all the way from Australia and one G-Forcer came from Canada!

We had many new GGs and G-Forcers. In the GG group, we've had two siblings who have galactosemia before but this was the first time we had three, 2 sisters and 1 brother—and all in one group! Old friendships were renewed and new friendships were made!

We started the conference with a BANG on Thursday night! Our hotel is the highest in Orlando—seriously! So, from the circular 27th floor with floor-to-ceiling windows, we could see fireworks from all the parks while we played board games. This was after the amazing Comedy & Magic show—no one will forget Joe tying the magician into the strait jacket—it's the only time he didn't have a camera in his hand!

We spent our mornings in sessions in small groups with the galactosemia professionals—where GGs and G-Forcers learned about the latest research, shared their experiences, and asked questions.

- New this year, were the breakout sessions where each GG and G-Forcer chose from one of four sessions that they were interested in—these were in-depth, discussions in small groups on Label Reading, Neurological Issues, Speech Issues, or Gastrointestinal Issues.
- World renowned dietician Laurie Bernstein, neuropsychologist Dr. Susan Waisbren, psychologists Dr. Roxanne Schreiber and Andrea Topper, along with special groups for the girls with the women doctors and guys with Dr. Berry, were among some of the other sessions the GGs and G-Forcers participated in.
- The GG and G-Force Biannual Lunch with the doctors and galactosemia professionals was another highlight—which not only provides GGs and G-Forcers with the opportunity to meet and find out firsthand about what these professionals are working on but it also provides the doctors and professionals the opportunity to learn more from the GGs and the experiences and issues they face.
- The Sibling Session with Dr. Roxanne Schreiber for the G-Force siblings was an excellent experience for each and every one—providing an opportunity for them to share their feelings and concerns about their sibling with Galactosemia. Dr. Roxanne said that she could see how much this group could really benefit from more opportunities like this at conferences.
- The No Whey Tropical Café gave everyone the opportunity to cook and taste new galactose-free recipes—Smoothies, and the big hit—Panini S'Mores!!

After lunch each day, the GGs and G-Forcers boarded buses to Downtown Disney where we went bowling at the really cool Splitsville Luxury Lanes one day, and then to Disney Quest with five floors of interactive games the next day.

The GGs and G-Forcers worked at the carnival to raise funds for the Paul Pruszynski Memorial Scholarship Fund on Friday night (the total amount of money collected: Over $800!!!) and then danced the night away on Saturday at the Teaming Together Farewell Social.

When the GGs and G-Forcers were not in sessions, they were seen at various venues in the hotel socializing with one

2014 Generation Gs

another—enjoying dinners together, hanging out at the pool, or game room. After all, the GG and G-Force programs are all about spending time with other young adults and teens with galactosemia. The diversity of the GGs and G-Forcers—many of whom are shy and have various difficulties—does not stop these phenomenal groups of teens and young adults from forming lifetime friendships and memories!

We thank the doctors and professionals who during their "free" time, made time for the GG and G-Force groups—you're involvement and dedication means the world to these teens and young adults—after all, you are their "avenue of information!"

And last but not least, none of this would have been possible without the continued commitment and devotion of the GG and G-Force parent volunteers, they have found getting to know these remarkable young adults and teens just as rewarding as I have. But without these volunteers there would be no GG or G-Force program—so a special thank you to each of you from Linda, Jeannine, and all the G-Forcers and Generation Gs!

So GGs and G-Forcers, get ready, we're already planning for Atlanta in 2016!

2014 G-Force

Round 5 Research Grant Announcement

The Galactosemia Foundation (GF) Research Team is happy to announce the fifth round of funding of its Research Grants Program. GF invites researchers to submit proposals for work that will increase our understanding of galactosemia and/or help families living with galactosemia. At least $75,000 in funding will be made available for the 5th round of grants. In order to allow for the award of multiple grants, applicants may request any amount less than or equal to $30,000, depending upon the project's scope.

Year of Award	Principal Investigator	Short Title of Research Grant	Grant
2007	Judith Fridovich-Keil	Toward Improved Intervention for Galactosemia	$50,000
2009	Susan Waisbren	Galactosemia: Review of Outcomes and Development of Guidelines for Management of Educational and Behavioral Challenges	$29,919
2009	Gerard Berry	Health and Psychosocial Outcomes in Adults with Classic Galactosemia	$30,000
2010	Sandy van Calcar,	Evaluation of Nutrition Recommendations for Classic Galactosemia	$27,000
2010	Susan Waisbren	The Brain and Neuro-functioning in Adults with Galactosemia.	$34,997
2010	Anna Marabotti	Computational Biology Strategy for the Development of Potential Drugs for People with Classical Galactosemia.	$41,500
2012	Gerard Berry	Modifier Genes and Epigenetic Effects in Galactosemia	$48,800
2012	Judith Fridovich-Keil	Toward improved long-term outcome in Classic Galactosemia	$40,361
2012	Kent Lai	Characterization of a New Mammalian Animal Model of Classic Galactosemia	$33,000
2012	Sandra van Calcar	Nutrition Management of Classic Galactosemia	$20,292

GF inaugurated the Research Grants Program in 2007 and awarded one grant in 2007, two grants in 2009, three grants in 2010, and four grants in 2012. The table below summarizes the projects that GF has funded to date. The window for submitting proposals for the fifth round of funding opens on Monday, December 1, 2014 and continues until Friday, January 30, 2015.

Visit http://galactosemia.org/Research.php for more information about the Research Grants Program and the current funding cycle.

THE AGERS GROUP IN ORLANDO

Kimberley Malyn

The AGERS group for older adults is a work in progress. The first group started in Texas and Orlando was our second time getting together.

We spent time getting to know one another, especially sharing and talking about our experiences. We enjoyed lunch together as a group and also cooking and tasting at the No Whey Tropical Cafe.

We attended some of the general session as well as breakout sessions that were of interest to us. The evening socials were a great opportunity to relax, talk to others with galactosemia as well as parents and families from all over the world—forming new friendships. It is always fun to be able to get to know other people who understand galactosemia and who have had some of the same types of experiences that we have. It isn't easy to keep in touch with one another since we are spread out across the country and world, so it is nice to have the opportunity to meet at the conferences.

We don't even have to be concerned about ingredient lists at the conferences because they always have safe foods. Having the opportunity to hear about the latest research from the various speakers helps us to understand more about ourselves. There are always new things to learn from the conferences, not just from the researchers but from other galactosemia families. As older adults, we are often approached by families as well as younger adults and teens—it means a lot to be able to share our stories, answer questions, and provide support—and know that we are in our own way helping these families and others with Galactosemia.

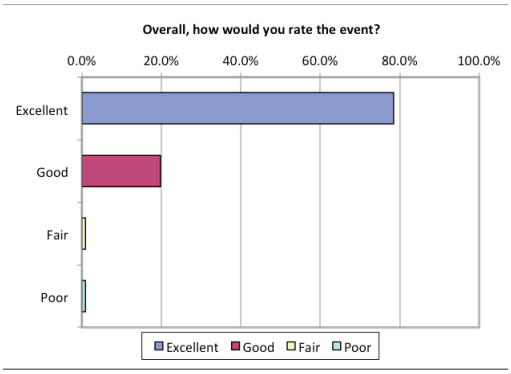
We are looking forward to 2016 in Atlanta. We have been discussing a new format for the group, where we will do more socializing among ourselves when not attending the general or breakout sessions. Dinners together in Atlanta, as well as some offsite touring-type activities are some of the things that we are looking into. Suggestions and ideas are welcome. Our goals include providing a fit for older adults and the opportunity to renew and make new friendships. As our group evolves, we would like to know what your expectations and goals are for AGERS. Please contact me with any suggestions, ideas, or questions at: agers@galactosemia.org.

Something New: AGERS Stories for the Newsletter

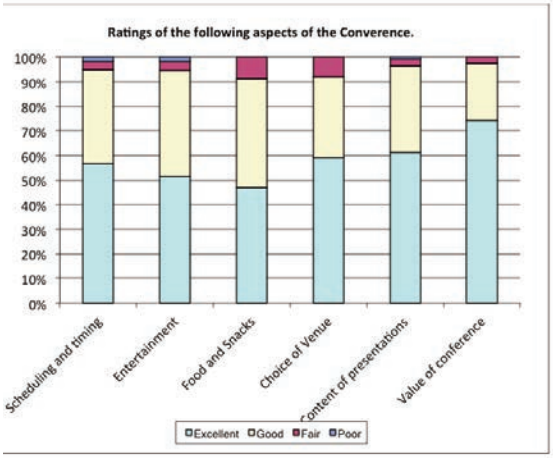
Share your stories—how and when were you diagnosed, your experiences, advice you have for other people with galactosemia and their families, or even just a particular story about something that you would like to share—ordering in restaurants, how you explain galactosemia to people who have never heard of it, and anything else. These accounts will be shared in upcoming newsletters. The hope is that your shared experiences will be helpful to others and provide insights into what it is like to have galactosemia. You can e-mail your stories to agers@galactosemia.org.

2014 CONFERENCE SURVEY SAYS . . .

Over five hundred people attended the Galactosemia Foundation conference in Orlando, Florida in July 2014. Here is what conference attendees had to say about the 2014 conference:



All aspects of the conference were rated as excellent or good by over 90% of respondents. Seventy four percent of respondents rated the conference as an excellent value. Only forty seven percent of respondents found the food and snacks to be excellent.



CANINE COMPANIONS FOR INDEPENDENCE (CCI)

Linda Manis

One of CCI's six facilities happens to be located in Orlando. Once we heard that the conference was going to be there, I approached them immediately about demonstrating the talents of these amazing dogs for our groups.

It was definitely one of the highlights for both the G-Forcers and GGs! The dogs not only showed off what they could do but also interacted with those who wanted to—shaking hands, delivering water bottles, dressing up, and more.

These special dogs (labs and golden retrievers) are bred in the CCI facility in California. There are six facilities across the country. CCI offers CCI dogs for children and adults who are in wheelchairs or have other physical needs, those who have difficulties with speech, learning, and/or socially, as well as those who have autism or autistic-type symptoms. These dogs can help those who have physical issues but for others, they are a companion, a friend, and more.

My son Adam, has had two CCI dogs—Wiskey, a black lab, is his current dog. Wiskey is Adam's responsibility—he feeds her, brushes her, walks her, and basically makes sure that she is well cared for. The best part is that Adam has a constant friend, and since Adam's best form of communication is by writing (e-mailing, texting, Facebooking), Wiskey has helped him to become more verbal—as he must give commands to Wisky to do various things—CCI dogs know more than 40 commands.

There is no charge for a CCI dog; however, it is a huge commitment. After an extensive application and interview process, once you are accepted into the program, the recipient and a family member must spend 2 weeks at one of the CCI facilities (they have special "dorms"), learning how to handle and care for these special dogs.

Seeing the GGs and G-Forcers with these dogs was a heartwarming experience their smiles told it all. If anyone is interested in learning more about CCI dogs, you can visit: www.cci.org

If you have any questions, feel free to contact me at: Lmscript1@aol.com.

What You Need

1/2 cup all-purpose flour
1/2 cup Hershey's unsweetened cocoa
1/4 tsp of baking powder
1/4 tsp salt
6 tbsp of Fleischmann's unsalted margarine
1 cup of sugar
2 large eggs
2 tsp of vanilla extract
Ghirardelli semisweet chocolate chips

Directions:

1. Preheat oven to 350 ° F (176.6 ° C). Line 8-inch square baking pan with foil. Grease foil.

2. On waxed paper, combine flour, cocoa, baking powder, and salt.

3. In 3-quart saucepan, melt dairy-free margarine over low heat. Remove saucepan from heat. With a rubber spatula, stir in sugar, then eggs (one at a time), and vanilla until well blended. Stir in flour mixture.

4. Spread batter in the prepared baking pan. sprinkle with chocolate chips. Bake for 18-20 minutes. Serve when cool.

Makes 16 brownies.

2015

Share your events with all The Galactosemia Foundation members. If you have a galactosemia event or fundraiser that you would like to have added to this calendar, please email newsletter@galactosemia.org

JANUARY	FEBRUARY	MARCH
	30th Anniversary of the Galactosemia Foundation/Parents of Galactosemic Children	
APRIL	**MAY**	**JUNE**
	30th Grant's Wish 5K Run, IL	12th & 13th GFSS Social, Decatur, TX
JULY	**AUGUST**	**SEPTEMBER**
		International Galactosemia Network annual researchers meeting, Lyon, France (during SSIEM congress)
OCTOBER	**NOVEMBER**	**DECEMBER**

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If you would like to sign up to receive this newsletter via email please go to our website: www.galactosemia.org

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