Galactosemia Foundation

Summer 2015

GALACTOSEMIA GAZETTE

PGC/GF TURNS 30!!!

INSIDE THIS ISSUE:

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

Galactosemia Foundation
Linked for Life.

In February 2015, the Galactosemia Foundation quietly observed its 30th anniversary. During that time the organization has assisted many families in connecting and advancing the conversation about galactosemia. The Gazette caught up with founding member Linda Manis, who reminded us how different the landscape was in the 1980s. "When my son Adam was diagnosed, the clinic gave us a 3/4 page handout!" stated Manis. "There was very little information available then. Library databases like LexisNexis had some details on other clinics, but the understanding of outcomes was poor."

The Original Board: (L-R) Rhonda Bauer (pregnant w/ second child, Susan), Gayle Dennis, Linda Manis, Christine Kovach, and Jane Wicks.

Manis began seeking out other families looking to learn from their experiences. Like today, privacy restrictions on health information made finding connections a challenge. As a workaround, nurses started providing Manis' phone number to other families, and she began to receive some calls. Initially, Manis invited five moms over for a lunch. "None of us had ever met another galactosemia family. We all felt like there had to be more of us." Manis began mailing her contact information to clinics around the country. "Initially calls were coming in around the clock from other families. Everyone wanted to do something."

Less than two years later, five women, Rhonda Bauer, Gayle Dennis, Christine Kovach, Jane Wicks, and Manis met in New York to discuss that "something." With Manis' mother serving as their attorney, the by-laws for Parents of Galactosemic Children (PGC) were drafted, the organization was incorporated, the first board was elected, and shortly thereafter PGC received 501(c) nonprofit status from the IRS. The board began working to open communication in the community. A newsletter was developed for families who had reached out and for metabolic clinics to share.

As the organization matured, it developed other forums to support the social and clinical needs of the community. "Early on there were two sparsely attended conferences in Ohio and in Texas," Manis recalls. "When Gayle [Dennis] took over as president, things progressed to the next level. It's incredible what the conference has become. It provides more for parents, and later we added the GG and G-Force programs for young adults and teens. At the conferences, our young people are so confident. They are like rock stars."

Manis also beamed when discussing the Foundation's connection with physicians. "We noticed that Adam had speech delays and heard other children were having similar outcomes. When we first asked the doctors, they claimed speech had no relationship to galactosemia. Later, a study was conducted, and it found there was a relationship." While supporting research was not the original focus of the Parents of Galactosemic Children, it has become a key strategy for the renamed Galactosemia Foundation. "[Guiding and funding research] was always a dream," said Manis. "It is great to have built such strong relationships with the experts. At the same time, there are still so many things we don't understand and I believe it is important for us to continue pushing in new directions."

Now: (L to R) Linda Manis, Gayle Dennis, Christine Kovach, and Rhonda Bauer.

Diet Details from Children's Hospital Colorado

By Laurie Bernstein, MS, RD, FADA, FAND

The diet for galactosemia is the lifesaving treatment for infants. With newborn screening and early initiation of the galactose-restricted diet, the life-threatening complications seen in untreated galactosemia can be avoided. These complications include poor feeding and weight gain, lethargy, jaundice, anemia, hypotonia, cataracts and a series of other signs that can lead to E. coli sepsis.

Because of the success of the diet in infancy, the standard recommendation has been that the galactose restricted diet should be maintained throughout life. However, the benefit of the strict diet has been questioned, given that galactose - 1 – phosphate concentrations in red blood cells do not return to normal even when the galactose-restricted diet is followed closely. In addition, despite having followed the diet, some individuals with galactosemia exhibit long-term complications such as language delay, speech defects, learning problems, cognitive and psychological impairment, tremor, ataxia, dystonia, and in females, premature ovarian insufficiency.

The Galactosemia Task Force (Sandy Van Calcar, Laurie Bernstein, Fran Rohr , Christine Scamen, Steven Yannicelli, Gerry Berry, and Denise Wilburn, parent representative from the Galactosemia Foundation) received a grant from the Galactosemia Foundation to investigate current knowledge and practice surrounding the diet for galactosemia. The task force searched the medical literature for information about diet and outcomes in galactosemia and food science literature for information about galactose content of foods. In addition, an online international survey was conducted asking questions from clinicians about infant feeding, diet transition for toddlers, calcium and vitamin D supplementation, restricted food and ingredients and lab monitoring. A paper was published with our findings in 2014 in Molecular Genetics and Metabolism. This is a summary of our findings:

Diet during infancy:

* Strong evidence for the benefit of a galactose restricted diet.
* Recommend soy based infant formula (powder, concentrate or ready to feed) or "elemental" formulas
* Discontinue breast milk or milk-based formulas
* Since soy formulas are not recommended in premature infants, an elemental formula is recommended in premature infants with galactosemia. There is little evidence that elemental formulas lower gal -1-p levels in infancy any faster than soy formulas.

Diet after infancy:

* AVOID Cow's milk and its derivatives:
  - o Casein
  - o Dry milk protein
  - o Dry milk solids
  - o Hydrolyzed whey protein
  - o Hydrolyzed casein protein
  - o Lactose
  - o Whey
  - o Lactalbumin

AVOID:

* Most dairy products need to be avoided (some are allowed)
* Avoid: milk, yogurt, ice cream, all cheeses except those noted

ALLOWED:

* All Fruits and vegetables

Including fruits, juices and vegetables in the diet has not shown to have any significant impact on red blood cell galactose-1-phosphate concentrations and /or urinary galactitol excretion.

* Allowed: Non-fermented soy based foods
* Allowed: Aged cheese: Gruyere, Swiss (Emmentaler), Jarlsburg, Tilsiter
* Allowed: Parmesan cheese (brick or grated) and sharp cheddar cheese that has been aged for more than 10 months
* Allowed: Calcium and Sodium Caseinates
* Allowed: All cocoa products except milk chocolate
* Allowed: Natural and artificial flavorings, all gums including carrageenan.

The task force recognizes that changes in diet recommendations may cause stress for some families. We recommend you discuss these recommendations with your clinic and dietitian.

There are diet education modules available that explain these new recommendations. http://www.ucdenver.edu/academics/colleges/medicalschool/departments/pediatrics/subs/genetics/clinical/IMDNutrition/Pages/IMDNutritionHome.aspx

If a family is uncomfortable with introducing new foods, it may be recommended to check the patient's red blood cell galactose -1- phosphate and /or urinary galactitol concentrations after introducing new foods. While these biomarkers are imperfect because they do not predict outcomes in galactosemia, they offer reassurance that the addition of new foods has not affected metabolic control.

Currently, the international community is also looking at developing international guidelines. Dr. Berry, Dr. Sandy Van Calcar and I are all a part of this effort. We will keep The GF community informed as the process continues.

ALLOWED:

- *All fruits and vegetables*
- *All non- fermented soy based foods*
- *Aged cheese: Gruyere, Swiss (Emmentaler), Jarlsburg, Tilsiter*
- *Parmesan cheese (brick or grated) and sharp cheddar cheese that has been aged for more than 10 months*
- *Calcium and sodium caseinates*
- *All cocoa products except milk chocolate*
- *Natural and artificial flavorings, all gums including carrageenan*

If you have questions or would like to contact Laurie Bernstein, please e-mail her at Laurie.Bernstein@childrenscolorado.org.

Scan this QR code for all the latest information.
Galactosemia Foundation

"Stronger Together" in Atlanta: 2016 Conference Plans Underway

By GF Volunteers
2016 GALACTOSEMIA
FOUNDATION CONFERENCE
JULY 14, 2016 – JULY 16, 2016
ATLANTA, GEORGIA

Planning for the 2016 Galactosemia Foundation is underway! Next year's event theme will be "Stronger Together." We are hoping for attendees from a broad geography to reinforce our worldwide stance for galactosemia research, education, and awareness.

On the logistics front, representatives from the board, the conference committee, and the research community are working with the Renaissance Atlanta Waverly Hotel & Convention Center to ensure a fun and "safe" experience. The hotel management and chef have been very understanding of our unique needs. Our group hotel rate will be $135/night +taxes for a standard room. The hotel has free internet and parking, two pools (indoor and outdoor), racquetball courts, spa and fitness center, jogging trails along the Chattahoochee River, and plenty of meeting space. Discounted airport transportation is also available to and from the hotel. Galactosemia "safe" lunches and snacks will be included with registration, while "safe" breakfast and dinner options will be available for purchase from the hotel restaurants. Corporate Kids Events will again be on-site to provide childcare at reasonable rates. On-line registration with cost and agenda details will be available early in 2016.

Atlanta, Georgia, is home to the 2016 conference, Coca-Cola, and countless other attractions that are sure to make the conference the best yet!

The education and social agendas for the event are also in development. Session topics will include the latest information on the social, behavioral, dietary, scholastic impacts of galactosemia, and updates on the ongoing research. Our goal is also to create opportunities for you, the registrants, to connect. We hope you can make new acquaintances, reconnect with old friends, access galactosemia resources, and network with the researchers and clinicians who are the experts on galactosemia. The Saturday night dance and photo booth will be back by popular demand as will the silent auction, the raffle, and the chocolate buffet of "safe" candies by the Illinois Nuts & Candy company. Offsite, Atlanta has lots to offer all ages including the Georgia Aquarium, the World of Coca Cola, and the Atlanta Braves baseball team. We are crafting programs for the G-Force, Generation G, and AGERS to experience the best of the region.

As in the past, our goal is to ensure the conference is available to anyone that is interested in participating . To make that possible, we need your help through finding sponsors, through fundraising, and through donations. If you have a corporate match program with your employer or have friends/family looking to assist, let them know their support can go directly to this impactful conference.

Feel free to reach out to treasurer lisa.spiro@galactosemia.org for help with any logistics of making a donation. If you have ideas for fundraising, feel free to connect with fundraising@galactosemia.org where the members of the Fundraising Team can assist you in maximizing your community impact. We are also always seeking sponsors, and if you have leads, please get in contact with conference@galactosemia.org. Finally, artists, we need your help too! If you are interested in having your creations considered for conference promotion, please submit them to conference@galactosemia.org by September 1st.

We truly are "Stronger Together."

Be sure to check out www.galactosemia.org for the latest conference updates and news, Foundation information, research updates, and fundraising opportunities!

"Our goal is also to create opportunities for you, the registrants, to connect. We hope you can make new acquaintances, reconnect with old friends, access galactosemia resources, and network with the researchers who are the experts on galactosemia."

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Woman With Classic Galactosemia Delivers Baby Girl!!!

Christin, Darrin Booth, and daughter Lucey Booth. Christin, who has classic galactosemia, delivered a healthy baby "Lucey" on April 1, 2014, in Nevada.

On April 1, 2014, Christin Lucey, a thirty-six-year-old woman with CG, delivered baby girl "Lucey Ann" weighing six lbs. nine oz. and measuring nineteen inches. Christin lives in Henderson, Nevada with husband Darrin. Christin (and her mom Paula Dellighattii) have participated periodically at PGC/GF events since her childhood. When Linda Manis learned of her exciting new arrival, she reached out to Christin to feature her story in the *Gazette*.

What day were you diagnosed with G?
I was diagnosed at six weeks by an ophthalmologist at St. Christopher's Hospital in Philadelphia who saw that I had cataracts. My liver was also inflamed, and I had been back and forth from home to hospital. They could not test me for galactosemia because I had had two full-body blood transfusions. My parents tested positive as carriers, and I later tested positive for CG.

What issues did you have (if any) growing up?
I was pretty shy, but got better as I got older. In primary school I was in some special ed. classes. I never liked being the only kid to have to leave my class to go to other classes. I motivated myself, and by high school I was in all regular classes. School was still difficult for me, especially math!

How old were you when you were told about POV?

When I was 9 years old, my mom had the "birds & the bees" talk with me and at the same time told me about POV. She said I may not be able to have children, but I could adopt if that were the case. At that time, the doctors were not sure if I would have ovarian dysfunction. I think my mom wanted me to be prepared for the worst.

How old were you when you started menstruating? Did you start with or without hormone replacement therapy (HRT)?

I started naturally when I was fifteen. I only menstruated once at fifteen though. It started again at sixteen, and at the time it was regular and normal. Then when I was twenty, it would stop and start—I would have it and then skip a month, and gradually it became more irregular. I had my hormones tested over the years, and they were fine. By my early thirties, my hormone levels started becoming high, which was not good—perimenopause. Then my hormone levels would go up and down. And I'd have my period for maybe six months. Then I'd start skipping again, and it would stop and then come back. My doctor basically said she didn't know what to do with me. She thought I could be in menopause.

My doctor wanted to put me on the pill, but it contained lactose so I didn't want to take it. I ended up using the NuvaRing for birth control. I tried the patch, which was fine for a few months, but then I started feeling weird. I felt dizzy, sick, and had headaches. I took off the patch, and then I was completely fine.

Was your pregnancy planned?

Not exactly. I went off of birth control because I was worried about being on it so long. I had been on and off before. My periods would be regular for about six months, and then they would start skipping a month again.

How did you find out you were pregnant?

My periods were pretty regular at the time, and I was a week late. I was at work and had gotten Subway for lunch. Anyone who knows me knows I LOVE pickles, but that day I could smell the sub (and pickles) from

"I didn't tell anyone because I wanted to wait and be sure. When I went to the doctor, she said that I was two months pregnant!"

the other side of my desk, and I just couldn't eat it—or for that matter anything! I was pretty nauseous. I decided to get a home pregnancy test, and it came back POSITIVE.

How did you feel about being pregnant?

I was scared but happy at the same time. I was in a new relationship, and even though we had talked about marriage and kids, I was worried about how my boyfriend would

CONTINUED...

handle it.

I didn't tell anyone because I wanted to wait and be sure. When I went to the doctor, she said that I was two months pregnant, and I had only missed one period!

I wasn't concerned about my galactosemia, but I was considered high risk because I was thirty-six. I didn't gain much weight, but the doctor wasn't too concerned. She said as long as I gained twenty lbs. it would be fine, and by the end I gained thirty lbs.!

Was Darrin tested for Galactosemia, or did you have an amniocentesis?

No, he wasn't tested, and I didn't have an amnio. We called Dr. Berry, who reminded us the baby would definitely be a carrier. She was tested when she was born, and of course is a carrier.

How was your delivery?

I delivered one week early. My water broke at 2 a.m. I was in labor for twenty hours. Two weeks before, I went into the hospital because they found out that Lucey was "butt down with her head and feet near the top of my waist." I had an external cephalic version to turn her. It took no more than sixty seconds to turn Lucey so her head was down, and luckily she stayed that way until the delivery.

I wasn't having contractions after my water broke, so then they gave me Pitocan to induce the contractions. That made the contractions so severe that I had to get an epidural. After the epidural, I was texting friends and family and rested until Lucey arrived. Everyone was in the delivery room when Lucey was born—Darrin, my Mom and Stepdad, and Darrin's Mom!

Was Lucey healthy?

Yes, she was fine. The only problem she's had is that she got sick and was gagging from my breast milk, so the doctor put her on a soy formula (Enfamil Prosobee). It made her gassy, so the pediatrician switched her to Nutramigen with Enflora. Now she drinks almond milk (just like me).

Other girls with Galactosemia are going to be so hopeful hearing your story. What would you like to say to them?

I'd like to tell other girls that being pregnant was a great time, and I loved it and it's something I had always wanted! But really it's in God's hands. so all I can say is it took a while and it took a lot of praying and hoping and going on and off the birth control to try to keep my periods. I didn't know if that would help, but I hoped for it and it is scary not knowing. But I had faith, and I just waited till I had found the right person. Darrin and Lucey have changed my life for the better, and I'm thankful everyday. I don't know why I kept my periods going, but we believe it was because my mom didn't drink milk at all while she was pregnant. That helped. There are other options also. There are many children in need of homes. Just keep your options open!

Christin pregnant with Lucey in Henderson, Nevada.

Would you be willing to talk to other girls with galactosemia and/or their moms? On phone? Facebook?

Yes, I'd love to.

Christin can be reached at:

E-mail: celucey@aol.com
Phone: 702-528-3046
Facebook: Christin Lucey (Henderson, NV)

Follow us on Pinterest for updates and information!

We know that there are other moms with Classic Galactosemia who have also had babies—We will be providing their stories in upcoming newsletters.

If you have had a baby and would like to share your story, please contact

Linda Manis
at
Lmscript1@aol.com

Like us on Facebook for information, events, and support!
Galactosemia Foundation

Boston-Bound Topper Tops the Class

By Keith Topper

With the commencement of the senior year of a high school student's life comes the ever tortuous and stressful college application process. Students across the globe will all be completing similar applications to many of the same post-secondary institutions. However, it is the college essay that will set all of these students apart from one another.

The essay is one of the most highly-debated portions of the entire application. Some people choose to express an important piece of their childhood—an event that has shaped them into the person that they are today. Other students will opt to write about a minor occurrence in their life, which taught them a lifelong lesson. The truth of the matter is that college admission representatives are always looking for fresh content in essays. Too many times students write a cliché story about how their sports injury sidelined them for their senior season. Or how the devastating loss of their grandfather from cancer caused them to begin to walk the annual Relay for Life event at their school. While these ideas may hit close to home, they would not set you apart from the rest of the crowd.

I decided to write my essay on how I endeavored on a journey to persuade food corporations into changing the ingredients of their food products. At the age of twelve, I began contacting large corporations such as Kellogg's and Nestlé, encouraging them to convert to allergy-friendly food products. I described galactosemia and how having the disorder has changed my day-to-day eating habits. Now I am much more aware of food labels and ingredients. In my letters, I proposed the idea of possibly eliminating milk ingredients and substituting them with soy-based ingredients or omitting the ingredient entirely. This would save companies money by not having to purchase an entire ingredient, which would result in profitability, and over the course of time, these margins would add up. Companies would also be benefitting by appealing to a larger customer base. Overall, it would be a win-win solution, where companies would be increasing profits, and customers would be increasing diet variety.

Not only was I most likely the only applicant to write such a unique, compelling story in my college essay, but I was also probably only one of a handful of applicants to mention galactosemia. My philosophy has always been to "milk the galactosemia." I try to get as much out of life as possible by using my unique story. I figured that the college essay would be the ideal place to "milk" my disorder by helping me to stand out from the crowd.

Keith Topper poses with his family and Dr. Berry at the Genetic Disorder Symposium at Harvard University. (L to R) Dr. Berry, Chris Topper, Keith Topper, Andrea Topper, Keith Topper Jr., Dr. McFaline.

Evidently, my college essay was a success. I am proud to announce that I have been accepted to the Questrom School of Business at Boston University with an annual $30K academic scholarship. Ever since I can remember, my family has taken my brother and me to see Dr. Berry. We have followed him as he moved to Philadelphia, Washington D.C., and most recently, Boston. Had it not been for the numerous trips my family has taken to see Dr. Berry in Boston, I may not have grown so fond of the city. Who knows if I would have even been able to travel to Boston had it not been for galactosemia?

If there is one thing that I could tell each individual student across the world, it would be to express yourself on your college essay. It is important that they discuss a topic that they are passionate about, a topic that they would be able to write a significant amount of detailed information on. The truth of the matter is that there is no "right or wrong" college essay topic so long as the student is passionate about his or her topic.

Linked for Life.

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 accomplishments.

Follow us on Twitter for updates and information!
@GalactosemiaFDN

Harvard Symposium Welcomes the Galactosemia Foundation

By Andrea Topper

The Galactosemia Foundation was invited to participate in a Genetic Disorder Symposium at Harvard University in Cambridge, MA on April 30, 2015. I was honored to represent our Foundation. Also on the Galactosemia panel was Dr. Gerard Berry and Christopher and Keith Topper, who both have Classic Galactosemia. The symposium was the culmination of the semester-long Genetic Disorder Project, which is an integral course requirement for students learning the principles of human genetics. The four disorders represented at the Symposium were Cri du Chat, Smith-Lemli-Opitz, Neurofibromatosis, and Galactosemia. Each disorder had a panel consisting of physicians, researchers, patients, and patient advocates.

Throughout the semester, students researched information about the effects of the disorder as well as the genetic and molecular underpinnings of the symptoms. After completing several different writing and presentation assignments about the disorders, the students attended the symposium. In many ways, this was the pivotal point of the course as an interactive discussion allowed students the opportunity to hear "real life experiences" from the individuals impacted by each disorder.

The Toppers and Dr. Berry participate in the panel discussion. (L to R) Dr. Berry, Andrea Topper, Chris Topper, Keith Topper.

During the panel discussion, Dr. Berry provided a brief scientific overview of galactosemia, an update of the collaboration with European countries, and updates on research he hopes to accomplish. Chris and Keith discussed the everyday challenges they face, which included academic struggles and triumphs. I addressed the mission of the Galactosemia Foundation, the goals of the various committees, information regarding the upcoming conference, and the imperative need for financial support for our Foundation. The panel discussion was followed by a very interactive, student-hosted question-and-answer session. After the symposium , many of the students stood in line to ask questions ranging from basic biology of the disorder to standard medical care, novel therapies and the social/family aspects of the disorder. After the symposium, we attended a reception where the students and panelists had the opportunity to chat in an informal setting.

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"In many ways, this was the pivotal point of the course as an interactive discussion allowed students the opportunity to hear "real life experiences" from the individuals impacted by each disorder."

Smile and Say "cheese" | Los Angeles Business Journal

By Charles Crumpley, Hannah Miet, and Melissah Yang
Monday, November 10, 2014

Six years ago, Jamie Siminoff's newborn son, Oliver, was diagnosed with a rare genetic condition that prevents him from metabolizing the sugar found in dairy products.

"When that happened, I started to be his partner in crime and not eat dairy," said the tech inventor, who founded Santa Monica video doorbell company Ring.

After discovering Kite Hill's almond milk-based cheese at Whole Foods, Siminoff contacted the Hayward company and asked if he could help with an investment. Siminoff, thirty-eight, put in $100,000.

"It's rare that somebody practically reaches out to a company and says, 'I love your products. I'd like to get involved. Can I help support you financially?'" said Kite Hill Chief Executive Matthew Sade.

It was a sound business decision for Siminoff, who likes to distribute his money in different industries in case the tech market crashes. But for the entrepreneur who mindfully invests in two projects a year, this one was personal.

"Your money is giving your child access to something they never had before," he said.

Siminoff; his wife, Erin; and his son, who has galactosemia, use Kite Hill's products to make cannelloni, ravioli, pizzas and even mozzarella-like sticks.

The now six-year-old gets to eat cheese at lunch every day, just like other children in his class.

"He'll eat containers of it," Siminoff said.

Source: http://labusinessjournal.com/news/2014/nov/10/smile-and-say-cheese/

Reprinted with the permission of the *Los Angeles Business Journal*.

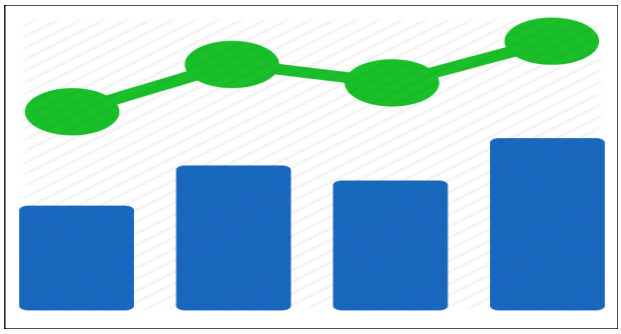
Follow us on Pinterest for updates and information!

"Without the support of the GF Community there would be NO GF Grant Program at all! If galactosemia research is important to you, please consider fundraising for the Galactosemia Foundation Research Fund today!"

Scan this QR code for all the latest information about the Galactosemia Foundation

Research Update—Round Five Research Grants Awarded!

By Denise Wilburn



The GF Research Team is happy to announce the 2015 GF Grant Award recipients. Three proposals totaling $90,375 were awarded in April 2015. The awards were given to Dr. Kent Lai, Dr. Judith Fridovich-Keil and Dr. Annet Bosch. Please see the "Grant Award Receipients" box for the details of each award.

2015 Researcher	GRANT AWARD RECIPIENTS Brief Description of Proposal	2015 Cost
Dr. Kent Lai	Mouse study - testing small molecule GALK1 inhibitors as therapy for CG	$49,500
Dr. Judith Fridovich-Keil	Is trace dietary galactose related to cognitive outcomes in CG?	$30,000
Dr. Annet Bosch	International collaborative meeting to establish international G guidelines	$10,875
Total		$90,375

In all, there were seven proposals submitted to the GF Research Team. The research team carefully chose proposals based on the GF research budget and the potential impact the proposal would have on the GF community. Four proposals were not funded due to the research accounts budget restraints. These proposals are outlined in the box "Unfunded Proposals."

	UNFUNDED PROPOSALS	
Researcher	**Brief Description of Proposal**	**Cost**
Dr. Sandy Van Calcar	20 test subjects. Extensive evaluation of nutrition and bone density	$27,675
Dr. Gerard Berry	Establishing a database of CG	$58,721
Dr. Manshu Tang	Mouse and cell study - pathophysiological mechanisms of accumulated gal-1-p in CG	$49,500
Dr. Patricia Jumbo-Lucioni	Identification of genes involved in Congenital Disorders of Glycosylation Using Animal Model	$44,000
Total Unfunded		$179,896

The GF Research Team thanks all those who submitted proposals, all the peer reviewers, and the GF Board for a successful round five. The GF team especially thanks all those who donated to the GF Research Fund. Without the support of the GF community, there would be NO GF Grant Program at all! If galactosemia research is important to you, please consider fundraising for the GF Research Fund today! We need to raise AT LEAST $100,000 by 2017 to fund round six. If you would like to help and don't know how, please contact us at research@galactosemia.org!

GAMA Announces Rylan Gelb Memorial Scholarship

By Denise Wilburn

The Galactosemia Association of Midwest America (GAMA) would like to announce The Rylan Gelb Memorial Scholarship.

GAMA established The Rylan Gelb Memorial Scholarship in 2014 after meeting a mom hero at the 2014 Grant's Wish fundraiser. Mrs. Stephanie Gelb bravely attended the event to support GAMA after she and husband, Hylton, lost their beloved baby Rylan from complications of galactosemia in December 2013.

Stephanie shared her story with GAMA and her determination to honor Rylan by making the world a better place, one act of kindness at a time, and encouraged others to do the same. Please visit rylangelb.blogspot.com for more information.

GAMA was so touched by the Gelbs and their story that we committed to join their cause of making the world a better place by establishing a scholarship in Rylan's name. The Rylan Gelb Memorial Scholarship will financially assist families with conference or evaluation expenses.

GAMA would like to help families pay for conference registration and offset travel expenses. The goal is to help families become more educated and make lifelong connections with others affected by galactosemia.

The second goal of the scholarship is to financially assist families so they can be evaluated by a medical professional specializing in galactosemia.

There are a few specialists around the country who have more experience in the treatment of galactosemia than others, and it can be very costly to get to one of these institutions to see a specialist.

GAMA would like to help families by offsetting their travel and direct care expenses.

Please visit http://galactosemiamidwest.com/rylan-gelb-memorial-scholarship/ for more information or to apply.

Rylan Gelb.

Dr. Berry Inducted into LaSalle College High School Hall of Fame

By Maureen Bell

On Sunday March 8, Dr. Gerard Berry was inducted into the LaSalle College High School Alumni Hall of Fame as part of a morning ceremony held in Wyndmoor, Pennsylvania, which is a suburb of Philadelphia.

A 1967 graduate of LaSalle High School, Dr. Berry was honored for his many years of service in the medical community and living the values of a LaSalle Explorer graduate, which are service, integrity, and respect. Dr. Berry in an acceptance speech spoke of how his years at LaSalle College High School fostered his years in medicine and dedication to serving and helping those in need, especially very young patients.

One of Dr. Berry's longtime friends and classmates Joseph Murphy introduced Dr. Berry and spoke of their friendship and his life-long commitment to the medical community.

Along with my husband and mother, I had the privilege of being in attendance at this special event. Also in attendance were Dr. Berry's wife (Nancy), son (Gerry), daughter-in-law, and granddaughter.

Dr. Gerard Berry with Maureen Bell after being inducted into the LaSalle College High School hall of fame.

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"The Rylan Gelb Memorial Scholarship will financially assist families with conference or evaluation expenses."

Do you have a story or information others in the galactosemia community would like to hear?

New ideas are always welcomed! Please e-mail newsletter @galactosemia.org with any ideas or suggestions.

AGERS, Generation Gs, and G-Forcers—You May Not Be Alone

By Linda Manis

Many years ago, speech and neurological issues (tremors) were deemed by the medical community to be unrelated to galactosemia. The efforts of parents to persuade medical professionals to take a closer look eventually sparked interest and curiosity, which led to research and eventually the recognition that there is indeed a connection to these issues and galactosemia. It is only the awareness of parents and family members that can influence researchers to consider galactosemia-related complications compared to issues that they may otherwise only "take a closer look" at researching.

Coordinators of the GG and G-Force programs, have seen that there are many different issues that teens and adults with galactosemia face—some have been proven to be related to galactosemia, and others are in question of being associated with galactosemia. One thing learned over the years, however, is that there is so much that families and researchers still don't know, especially about teens and adults.

During the past couple of years, Jeannine Quam's son Tyler (21) has experienced a number of physical issues including chronic pancreatitis, a rare nasal tumor identified as Juvenile Nasal Angiofibroma, nonepileptic seizures, and severe reactions to Propofol, a general anaesthetic.

Recently, Linda Manis' son Adam (31) experienced a severe reaction to a medication combination he was taking to help control his tremors. In addition to a seizure, he also experienced a number of mental health-type issues. Fortunately, after a couple of months of ups and downs, he's finally doing better. It is known that there are other adults, including Ben Huber [39] from Michigan, who have had similar issues although not always triggered by medication.

Adam, Tyler, and Ben may not be alone and there are possibly other teens/adults with galactosemia who have had the same or similar issues. These could be related to galactosemia or not, and this is why research is needed.

The first steps are to see what other teens and adults have experienced or are experiencing and to see what treatments and/or medications were successful or unsuccessful. It is the hope that teens, adults, and parents will share their experiences with us. Please respond if anyone is known to have experienced any of the following complications:

- Crohn's Disease
- Colitis
- Constipation (researchers have shown an interest)
- Pancreatitis
- Juvenile Nasal Angiofibroma / Tumors
- Seizures (Seizure Disorder)—If So, What Type of Seizures
- Severe Reaction to Medication or Medication Combinations
- Sadness / Depression
- Nervousness / Anxiety
- Paranoia
- Hallucinations or Delusions
- Anxiety
- Other – If there is anything that you have experienced that does not include more commonly known galactosemia complications (speech and learning issues, tremors), it's important for us to know about any of these too.

Susan Huber, a mental health nurse and Ben Huber's mother, states, "I believe that "putting it all on the table" just the way we see it and calling out specific mental health issues such as paranoia is how we get to the root of the problems. Many people can discuss mental health in the context of depression and anxiety, but when it turns to delusions and hallucinations, they often withdraw from the conversation. If we lead openly with our adult children we are more apt to get an honest discussion going."

The plan is to assemble this information and then make it known to the Galactosemia Foundation and to researchers. The hope is that by gathering this information, interest from researchers could possibly result in more rigorous studies. The information will provide an overview of the number of adults who responded and a list of the complications encountered.

Responses can also help the G-Force, GG, and AGERS programs along with the Galactosemia Foundation to determine whether there is a need for speakers who can provide more information on these topics at the upcoming 2016 conference in Atlanta.

Tyler, Ben, and Adam should be commended on their bravery to come forward and share their experiences with the Galactosemia Foundation. As Tyler put it, "I want to learn more and help others affected by galactosemia."

Having conversations regarding what is occurring in the lives of adults with galactosemia will lead to clarity and a better understanding of life as a person living with galactosemia. It is the hope that this information will help unite galactosemia families and provide medical professionals with the motivation to explore further research opportunities. "Connecting the dots" will hopefully help all people living with galactosemia have a bright future!

Please send any correspondence to:

Generation G Coordinator: Linda Manis: Lmscript1@aol.com / Call or Text: 954-610-3739
G-Force Coordinator: Jeannine Quam: jmquam@edenpr.org / Call or Text: 612-232-0171

Please remember to include age, sex, issues experienced, duration, longevity, severity, general health concerns. Thank you for your participation. Note that all information gathered will be held confidential. It is only through sharing information that progress can be made.

DOES RIGOR OF DIETARY GALACTOSE RESTRICTION IN EARLY, MIDDLE, OR LATE CHILDHOOD ASSOCIATE WITH DEVELOPMENTAL OUTCOMES IN CLASSIC GALACTOSEMIA?

NO ONE KNOWS....

BUT WITH YOUR HELP WE CAN FIND OUT!

If you have classic galactosemia, please consider volunteering for our research study (Emory IRB00024933, PI: JL Fridovich-Keil, currently funded by the Galactosemia Foundation). We need both child and adult volunteers.

Participation in this important study is easy, free, and requires no travel or extra needle stick.

What would we ask you to do?

- Complete an informed consent process.
- Complete an online or phone survey about diet history and current diet.
- Help us get access to your current or historical school records including standardized test results.
- Give us a small sample of your blood collected in conjunction with a normal clinical blood draw so there is no extra needle stick. If you do not have clinical blood draws we will ask for a sample of saliva instead.
- This study also has other parts that you might want to participate in but you would be free to say no to any parts you did not want to do.

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!

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.

BRONZE LEVEL - Raise $750.00 – $2,499.99*	**SILVER LEVEL - Raise $2,500.00 – $4,999.99***
• One Conference registration fee waived • Recognized on conference handout • Entered into a drawing for an I-pad mini	• Two conference registration fees waived • Recognized in general session and conference handout • Entered into a drawing for an I-pad mini **1st Place**** - Reimbursement for up to two nights stay in the conference hotel***
GOLD LEVEL - Raise $5,000.00 – $9,999.99*	**PLATINUM LEVEL - Raise $10,000.00 or more***
• 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*** **1st Place**** - Reimbursement for one coach round trip airfare ticket***	• 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: Galatosemia 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