Spring/Summer 2013

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

Orlando 2014 – Detailed Planning Has Started!

I know that the Galactosemia Foundation Conference in July of 2014 sounds like a long time from now, but your Galactosemia Foundation Board has already met to begin planning the conference! On Saturday and Sunday, February 9th and 10th; the Galactosemia Board met in Orlando to talk with the hotel staff and build the plan to ensure the conference is a success.

The board started with a review of the survey information from the past conference and identified what-worked and what did not work at the 2012 Conference in Dallas. Many successful things that worked in Dallas will be carried forward into the Orlando conference, including onsite child care, a conference incentive program and a list of topics.

Most importantly the board spent time on the goals and objectives and theme for this conference. The theme will be centered on: Teaming Together for a Bright Future. This theme is a recognition that we all come together to achieve a common goal (teaming together) to provide a bright future for all of us and our children.

The board spent most of the weekend's time:

1. Outlining the timeline for the conference days: Thursday night, Friday and Saturday including meals, speakers and entertainment activities
2. Discussing current and projected conference budget
3. Reviewing the conference incentive program guidelines
4. Brainstorming speakers and topics
5. Developing a complete communication plan to make sure we engage every media possible to make the 2014 conference the biggest and best yet.
6. Planning for vendors and sponsors to support the conference

Since this was the initial planning meeting, the board and conference committee have planned monthly conference calls and plans to meet at least one more time to continue to plan for an exciting 2014 conference.

Much time goes into planning for this event so please, mark your calendars now.

If you have any questions on the upcoming conference, please go to the website: www.galactosemia.org or post on the facebook page https://www.facebook.com/Galactosemia.Foundation or submit your question to conference@galactosemia.org.

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.

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

The Galactosemia Foundation
P.O. Box 2401
Mandeville, LA 70470-2401

Taking a look at technology...

In our world today, everyday technology consists of laptops, notebooks, smartphones, tablets, texting, and a variety of social media outlets. The internet is a wealth of information with millions of resources right at our fingertips.

How can technology help individuals that might be struggling with the complications of galactosemia? What about individuals that find typing a challenge? Are there alternate ways to compose a message other than typing? Is there a solution to address difficulties with reading books, websites, or information on a computer? Are there tools to help? What about individuals with vision issues? Can we offer alternatives to see and access materials? For parents, how do you help support your child when they might be struggling with academic subjects?

The answer to all of the questions above is yes. These issues are shared by many individuals, regardless of the disability, diagnosis or disorder. Assistive technology includes a wide array of tools and resources to help individuals with disabilities. Some tools are free or low-cost, and available to everyone. Other tools have specific costs and materials are selected to meet the individual need a student or adult (e.g., someone who is non-verbal might require a device to speak for them).

One article on technology cannot begin to explain the thousands of features and tools available for individuals. The information below includes some great resources regarding technology and getting started:

Did you know that accessibility features are built directly into your computer, smartphone or tablet ? Some devices offer a robust array of features, others include a minimal amount.

Some commonly used accessibility features might include:

- voice recognition (also called speech to text; you speak into device and words are printed on the screen)
- inverted colors (changes the colors on device; making it easier to see for individuals with low vision)
- magnifier (enlarges the text on the screen)
- screen reader (Windows) or Voice over (Mac) will read information on screen aloud
- closed captioning
- keyboard and mouse modifications (e.g., onscreen keyboard, enlarged mouse pointer, sticky keys)
- Guided Access (specific to Apple mobile devices; this allows the user to lock into apps, restrict parts of apps)

For more information regarding accessibility features,check out a few of these websites:

- http://www.apple.com/accessibility/
- http://www.microsoft.com/enable/products/windows7/
- http://www.microsoft.com/enable/products/windows8/

Check out some of these great free or low-cost tools across academic areas. There are so great tools this is just a sampling! Here are a few great resources:

Reading/Writing:

- Tarheel Reader http://tarheelreader.org is an online website with a variety of easy-to read stories; plus you can make your own! Each has speech-to-text and the stories are switch accessible.
- Cast UDL Book Builder http://bookbuilder.cast.org is an online bank of stories, with a variety of levels (K-12), The stories have speech-to text and uses can sign up to make their own stories.
- Inkless Tales http://www.inklesstales.com includes online stories, poems (lower elementary) with Dolch words (also has some comprehension/predicting questions on website).
- Spelling City http://www.spellingcity.com is an online resource to practice spelling (K-12), also helps to develop reading and writing skills!
- Kerpoof http://www.kerpoof.com is an online website geared for K-8 students that integrates pictures and creativity (pictures, videos, card-making) into spelling and writing practice.
- Bookshare https://www.bookshare.org is an online accessible library of thousands of books for individuals with print disabilities. Most schools have free memberships, individual memberships are $50 per year. Individuals must qualify for Bookshare (with visual, learning, or physical disability).
- Some great reading/writing apps for the iPad include: Notability (1.99), Clicker Sentences ($20.00) or Clicker Docs ($25.00), App Writer US ($29.99), Paper Port Notes (free), Word Wizard (1.99), and Story Book Creator (FREE).

Math:

- iSolve it puzzles http://isolveit.cast.org/home are online math activities for middle school students to help develop thinking and reasoning skills.
- Time for time is a great website for learning concepts of time http://www.time-for-time.com
- Math Playground is a great website for Math Games http://www.mathplayground.com/games.html
- For the iPad, check out: Math Girl, Math Ninja, Mathmateer, Splash math, Motion math, Montessori Numbers, Space Math, math apps by McGraw Hill (often go on sale).

Science/Social Studies:

- CAST science writer (http://sciencewriter.cast.org/welcome) makes writing science reports easier! It is geared for ages 6-10. It also integrates text to speech.
- The Concord Consortium http://concord.org is an online website that includes activities for science (and math) http://concord.org. It is geared towards upper grades with some activities for elementary school.
- iPad Apps that target science and social studies: Stack the States, Stack the Countries, Presidents vs. Aliens, The Elements, Science 360 for iPad (free).

Typing:

- For typing games, check out http://www.learninggamesforkids.com/keyboarding_games.html
- Slime Kids has a variety of online typing activities http://www.slimekids.com/games/typing-games/
- Typeracer targets increasing typing speed http://play.typeracer.com
- For the iPad, check out: tap typing, ghost type.

A few tidbits of information about technology in education:

One of the "up and coming" topics in education is a term called Universal Design for Learning (UDL). As we know, not all individuals learn the same way--the UDL model proposes that classrooms have a variety of tools and materials (often free or low cost) to meet the learning styles of all students. For more information on UDL, check these websites out:

- http://www.cast.org/udl/
- https://sites.google.com/site/nhinstitutes/21st-century-learner/using-udl-to-support-a-diverse-classroom
- http://www.cpacinc.org/wp-content/uploads/2009/12/ParentsGuidetoUDL.pdf

If your child has an IEP, all teams must "consider" assistive technology for students. For more information regarding AT and the decision-making process for AT (for students with an IEP), here are some great links:

- http://www2.ed.gov/policy/gen/guid/assistivetech.html
- http://aim.cast.org/learn/stakeholder_focused/parents_students
- http://www.joyzabala.com

Have a specific assistive technology question? Want to share the technology that someone with Galactosemia finds beneficial? I would love to hear about it! I am mom to Ava Dittmann (age 2 1/2) who has Classic Galactosemia. She loves technology!

Please email me at kellydittmann@me.com!

Galactosemia Foundation
Treasurer's Report
2/23/13

Net Financial Position

Our net financial position as of 2/23/13
Is as follows:

Assets	
General Account	$ 28,085.20
Research Account	$ 144,475.11

Total Assets	$ 172,560.31
Liabilities	($ 75,000.00) (2013 Research Grants)
Net Position	$ 97,560.31
	=========

Key Activities

- 2014 Conference Incentive Program: The 2014 Conference Incentive Program is open. This is an easy way to help offset the cost of attending the Orlando conference. All you have to do is get your family, friends and co-workers, even friends of friends to donate money to the 2014 conference and you could earn free registration, free hotel room nights, and even free airfare. The key is all monies donated have to be directed to the 2014 conference to be eligible. Please look for a full breakdown and details of the conference incentive program in this newsletter or contact me at treasurer@galactosemia.org.

- Fundraising: There are tons of simple easy ways to raise money, from lemonade stands to golf tournaments; our Fundraising Committee has put together some helpful information that's a simple email away at fundraising@galactosemia.org. Remember the Galactosemia Foundation is a Non-profit 501(3)(c) so donations are tax deductible.

Round 4 GF Research Grant Awards Announced

GF received six strong proposals for Round 4 of GF's research grant program. Each proposal was reviewed by the research team lead to ensure it included all required information prior to being officially accepted. Fourteen peer reviewers were recruited and two were assigned to each proposal by the research team lead. Each peer reviewer reviewed one proposal using GF proposal guidelines and a provided scoring rubric. In parallel, the research team members each reviewed all proposals and evaluated their usefulness to the GF community and their plan for sharing any information generated by the research or study. A recommendation was made by the Research Team to the Board of Directors to fund four of the proposals at a total cost of $142,453, leaving approximately $8,000 left in the research fund. The GF Board of Directors approved the research team's recommendation. The four funded research grants are summarized below:

Principle Investigator	Project Short Title
Dr. Gerard Berry	Modifier Genes and Epigenetic Effects in Galactosemia
Dr. Judith L. Fridovich-Keil	Toward improved long-term outcome in classic galactosemia
Dr. Kent Lai	Characterization of a New Mammalian Animal Model of Classic Galactosemia
Dr. Sandra van Calcar	Nutrition Management of Classic Galactosemia

Each of the research grants will be awarded for a duration of one year. Each of the research grant recipients will be asked to write an interim "lay" summary of their research after six months, which will be included in a future GF newsletter and posted on the GF Research Web page. In addition, each research group will write a final report at the completion of their grant and these reports will be posted on the GF Research web page. In addition, we have asked the lead of each of the research groups to consider presenting the results of their work at the 2014 GF conference in Orlando.
Research Team members include Dan Lambert (Lead), Brian Mannix, Beatrice Ortego, Jo Beth Southard, Christy Johnson, Denise Wilburn, Kristine Saylor, Christine Winey, Mark Winey, and Kayla Costner. Contact the research team via e-mail at research@galactosemia.org.

Goodsearch Update

Our GoodSearch (www.goodsearch.com) fundraising contest officially ended on February 28th.

Congratulations to our top 3 supporters during the contest period:

Christina Dell
Amy Hunt
Kenn Gall

Please contact vicepresident@galactosemia.org if you haven't already claimed your fabulous prizes!

Thank you to those that became Galactosemia Foundation supporters at GoodSearch. Even though the contest is over, please become or continue to be a supporter and help us earn donations every time you search the net and shop online!

More Ways to Raise Money via www.goodsearch.com
Goodsearch recently launched two new ways to earn donations: surveys and offers. Now Goodsearch offers browser "add-ons" to make using GoodSearch easier. These are in addition to searching the internet, shopping online, and dining. See http://www.goodsearch.com/faq to learn more about all of the features of GoodSearch and how it all works.

Tips and Tricks for Effectively Using GoodSearch

1. Always make sure that you are logged in and supporting Galactosemia Foundation – otherwise the foundation won't get any donations from your searches.
2. Set GoodSearch as your home page on all of your computers and devices that you use to access the Internet at home and at work. See http://www.goodsearch.com/getinvolved.aspx for instructions on how to set your homepage.
3. If you use Internet Explorer, Firefox, or Chrome, install the "GoodSearch App" to automatically make GoodSearch your default search engine. If you use Firefox, the App will add a GoodSearch searchbox to your toolbar.
4. If you use Internet Explorer or Firefox, install the "GoodShop Toolbar" to add a GoodSearch search box to the top of your browser and to automatically get credit for any purchases from a GoodShop merchant without having to click through the GoodShop page.
5. If you use Chrome or Safari, install the "Goodshop To-Go Button" - which is similar to the "GoodShop Toolbar" but it doesn't add the search box.

You cannot spell Fundraise without FUN!!

Our family, The Saylor Family, was inspired to fundraise at our first conference by Mark and Jo Beth Southard. I sat in the audience and listened to the passion they had to build awareness and raise money for research for galactosemia. My wife Kristine and I said we need to do something! We thought even if research does not help our son Jake we need to help the future generations of galactosemics. We started with a small golf outing and over the years it has grown and grown. Considering we are originally from Cleveland, Ohio and have no family or deep roots in Richmond, VA we did not know how big we could make our golf outing. But we found when you give people a chance to be generous, they will amaze you!

We need more families to "just do it" and start fundraising. Big or small, everything helps. We cannot fund research and have conferences without the outstanding work of all the families who dedicate themselves to fundraise. The first thing is picking something you like to do (golf, run, cook, etc.) You can make your event as big or as small as what fits you. You do not need experience, there are plenty of people to help you get started.

Need some ideas?

- 5 K run
- Golf outing
- Dinner/Auction
- Costume party
- Raffle, Reverse Raffle
- Bowl-a-thon
- Video-game-a-thon
- Bake sale, Lemonade stand, etc.
- Selling your old things and donating the money.
- Charity night at a local restaurant (Many will give you 10% of bill for people who go to support your charity.)

There are many more ways to fundraise. All you have to do is find out a way to raise money based on what you love to do. Once you outline what you want to do, reach out to the fundraising@galactosemia.org or myself and we would be glad to help you get started. Consider an event that is sustainable year to year. Believe me it does get easier and easier every year.

Coin Box Fundraiser

The fundraising team is sponsoring a summer coin box fundraiser. How many of you (yes YOU, the person reading this newsletter) would be willing to fill a little box with change? You can do that, RIGHT? How many of you think your friends and family would also fill a little box? You're all shaking your head YES….right? If we can collect the money that will fill 500 little boxes we will raise $10,000+ for the Galactosemia Foundation.

Are you with me?

Here's how it works. We would like for you to think about how many friends and family members would fill a 3" box for you. E-mail Denise Wilburn at fundraising@galactosemia.org or Michelle Fowler at president@galactosemia.org to request the amount of boxes you can fill. We will happily send you boxes with the agreement you will do your best to have them filled, cash the coins in at your local bank and send a check for the full amount to GF.** Together we could raise enough money to fund part of a research study or support the next GF conference. If you have any questions please e-mail Denise (fundraising@galactosemia.org).

* You can register the coin box fundraiser as your fundraiser for the contest!

**Please be aware it costs money to ship the boxes. Please only request what you will fill and return to GF.

Announcing The First GF Fund Raising Contest!

The fundraising team is proud to announce a new FUNDRAISING CONTEST! We will be awarding prizes for fundraisers held in the months of June, July, and August of 2013! We will offer prizes for the most money raised, the most creative, and the best first time fundraiser.* We have some awesome prizes including an I-PAD Mini, a Kindle Fire, Galactosemia Foundation goods, restaurant gift certificates, and more! Just register at galactosemia.org to get started. All fundraisers must be registered BEFORE they occur to be eligible for the contest.**

Never organized a fundraiser before? It's okay! Check out the article in this newsletter or our website for ideas or e-mail Denise Wilburn at fundraising@galactosemia.org for assistance.

*First time fundraiser names will be drawn at random to receive the prize, we hope to have many!

**Donations must be received and cleared by our treasurer no later than September 15th to be eligible for the contest.

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.

Are You Talking To Me?

When it comes to fundraising many of us don't think we are equipped nor have the time. We think it is a great idea, but when it comes to ACTUALLY doing it….we think its best left to someone else. The fundraising team would like you to know ANYONE can fundraise….INCLUDING YOU! A fundraiser does not have to be huge or take months to plan. A fundraiser that raises $50 is as appreciated as one that raises $5000. If every one of our families raised $50 this year we would have $20,000 to use for support, education and research. The GF is in need of replenishing both the general and research funds! We are planning an awesome conference for 2014 and need funds to support it. We also just funded research projects and need to replenish our research fund! If you appreciate a newsletter, our website and conference please consider giving to the general fund. If you would like to see new research or encourage new researchers to take an interest in studying galactosemia PLEASE give to the research fund!

Here are some ways you can fundraise:

1. Keep a jar marked "Galactosemia Foundation Donations" in a visible area of your home. Put all your change in the jar on a regular basis and when guests ask what it's for you can explain…maybe they will drop a quarter or two!
2. Send out a letter to family and friends letting them know you are raising money for this great cause during the month of June (or whatever month) and ask them to make a donation. Remind your family donations are tax deductible and can be made by debit or credit card through our website galactosemia.org.
3. On your next birthday ask for donations instead of gifts.
4. Hold a backyard BBQ where you provide the BBQ and ask friends to bring a donation.
5. Throw a party to celebrate anything (your blessings, health, sunshine, the Super Bowl, NASCAR, that grass grows, whatever) and collect a donation at the door.
6. Turn your hobby/talent into a fundraiser. Exchange your talent for a donation.
7. Check with your local race track, concert venue, Dairy Queen or local restaurant: many businesses will give you a cut of profit if you work a few hours at their location.
8. Plan a 5K, poker run, silent auction or have a booth at a local event.
9. Plan a bake sale around another event (like a 4th of July parade or a Summer Fest)
10. Participate in the Coin Box Fundraiser described in this newsletter.

The fundraising team has been working hard to encourage EVERYONE to take an interest in raising money for research and education. We have 2 great opportunities planned for the summer 2013. First we are sponsoring a fundraising contest with great prizes. You'll have a chance to win an I-Pad Mini, a Kindle Fire, gift certificates and cool GF merchandise. Register your fundraiser on the GAMA website (galactosemiamidwest.com) and get planning! We are also sponsoring a GF Coin Box Fundraiser. You and your family can fill provided coin boxes with change. Cash in the change at your local bank and mail a check to GF. It's that simple! Check out the other fundraising articles in this newsletter for more details.

The First Ever Galactosemia Foundation Golf Tournament!!!

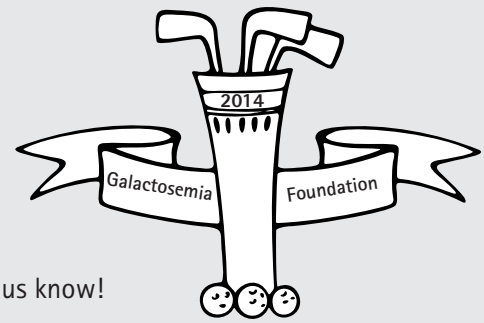
All you golfers (and non-golfers)—we are planning a fund-raising Golf Tournament in Orlando!

We are putting together details but do need to hear from you as to whether there is a significant interest!

When: Thursday, July 17th (Registration Begins Thursday evening)

Time: Golf in the morning and then lunch

Cost: Between $100–$150 per person (will include green fees and lunch)



Sponsorship opportunities will also be available.

If you are interested in participating in the Golf Tournament, it is important that you let us know!

Please contact Linda Manis or Joe Leggio! Linda Manis (Leggio): Lmscript1@aol.com Joe Leggio: Joe.Leggio@aol.com!

Suggested Reading Literature:

The Out-of-Sync Child
by Carol Stock Kranowitz, MA
"Recognizing and coping with Sensory Integration Dysfunction." This is a great book, written so that parents and professionals can understand what is happening, and what can be done to help.

Practical Procedures for Children with Language Disorders
by Donald Nelson, PhD
This is a wonderful book for parents, speech therapists, grandparents and professionals. It has great tips and ideas. The book is written so everyone can understand the depth of language disorders and that the correct therapy can help.

So Stick a Geranium in Your Hat and Be Happy!
by Barbara Johnson
"Pain is inevitable but misery is optional." This is a fun read, like a Christian Erma Bombeck. She has written many others like "Where do Mothers go to resign?"
The Brand New Kid: by Katie Couric
This is a great kid book about people that are "different."

PLEASE SUBMIT SUGGESTIONS OF LITERATURE, WEBSITES, OR APPLICATIONS TO RTABB7@comcast.net to be published in an upcoming newsletter or the website.

***As Galactosemia Foundation, we are only passing on information when referencing literature, websites, and applications. It is your judgment and knowledge that needs to make the decision on whether it is fitting for your family to be beneficial or not.

What You Need
1/4 Cup cold soy milk or rice milk
1 pkg. (3.9 oz.) (Best Choice is a possibility)Chocolate Instant Pudding
36 OREO cookies, finely crushed (about 3 cups)
12 oz. BAKER'S Semi-Sweet Chocolate or chocolate chip of choice, melted

Make It
STIR milk and pudding mix in medium bowl with large spoon just until pudding mix is moistened. Immediately stir in cookie crumbs; mix well.

SHAPE into 42 (1-inch) balls. Freeze 10 min. Dip in semi-sweet chocolate; place in single layer in shallow waxed paper-lined pan.

REFRIGERATE 20 min. or until firm.

Participate in the Recipe Contest

Please submit **Main dish recipes** for upcoming newsletters and the kitchen wizards will choose which one is the hit to be published in the next issues.

1) You have one month after your newsletter is received to submit the recipe to rtabb7@COMCAST.NET.
2) MUST BE GALACTOSEMIA FRIENDLY.

Outreach Team Spring 2013 Update:

We are always busy welcoming new families; some of our newest families have been from San Francisco, Indiana, North Carolina and Madrid! However, we couldn't do so without the help of many of you! Thank you so much for sharing your contact information, experiences and time with the new and old families these past few months. Nearly every one of us remembers that first email or first phone call from a contact that helped us through the initial few weeks and months. Volunteer contacts, you're making a huge difference! If you are interested in volunteering your contact information and sharing your time with new families as they pop up in your area, contact us at Robyn & Jeremy Meek, meekfamily1@gmail.com.

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One great benefit of Outreach is our ability to connect people who need to share information together. Recently we have had two families with specific needs that could use a little more help. If you have any experience with endocrinologists & growth hormone therapy, or epilepsy/seizures & tremors, please contact the following families listed below:

EDOCRINOLOGISTS & GROWTH HORMONE THERAPY: contact Jessy: jldershem@gmail.com

EPILEPSY/SEIZURES & TREMORS: contact Kelly: jkdittmann@yahoo.com

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Outreach is thrilled to have two new enthusiastic team members helping out!

Tara Tanella of New York & Tara McCoy of Kentucky!
Meet Tara Tanella!
She can be reached at: ttanella@gmail.com
My name is Tara Tanella, and my son, Joseph, was born on February 8, 2011 with Classic Galactosemia. We were notified by our NY State assigned "galactosemia" hospital (Stony Brook) to rush Joseph to the hospital on day 3 of his life, after being home for less than 24 hours. Luckily, we live in Whitestone, New York which has newborn screening results within 5 days. As first time parents, my husband and I were scared and in disbelief. The scariest part was that we never heard of galactosemia. We didn't know what to expect, or what this even meant for our baby. Joseph became very sick and was fighting for his life. Joseph received numerous treatments to help increase his organ functionality, which included bili lights for jaundice, vitamin K shots and a white platelet transfusion. Due to the care of Joseph's amazing doctors and the persistent strength of our new baby boy, we received the news that we could take our little love home on Valentine's Day! Without the support of our family and our galactosemic families, we would not be where we are today. Joseph is currently under the care of the Metabolic/Genetic teams at Mt. Sinai (Dr. Melissa Wasserstein) and Boston Children's Hospital (Dr. Gerard Berry). Joseph is a happy, sociable two year old with the greatest personality. He loves to laugh, sing, read his books, climb, do crafts and play with other children. He is currently hitting all milestones of development and, most importantly, always smiling and full of life. He is also preparing to be a big brother in October. Joseph brings so much love and joy to us and to everyone he meets. We're very blessed to have him as our son.

Meet Tara McCoy!
She can be reached at: Tara.McBride@insightbb.com
My name is Tara McCoy and I have a daughter, Brinley, who was born last summer with Classical Galactosemia. We live in Louisville, KY and were surprised to find that there was only one other family that has an adolescent daughter with Classical Galactosemia in the area. We found them through a friend of a friend who saw our posts on Facebook as we were spending days at the children's hospital to find out why Brinley was so jaundiced and just couldn't seem to gain weight. It was so scary at first to hear all of the horrible things that could happen if she did in fact galactosemia. It wasn't until we met up with our new friends who have a perfectly healthy and outgoing daughter living with galactosemia, that I began to relax a little and enjoy my sweet baby girl instead of worrying. They gave us tips about where we could eat when she gets older and how to prepare foods in a safe way. She assured me that Brinley would live a normal life just like every other kid, I would just have to be a little more prepared and cautious. This is why I have a passion to help others who are in the same situation as I was last summer: scared, overwhelmed, confused and dealing with so many other emotions. I am hoping to help others through those first few months by helping them connect with other people in their area and with the best medical care. We are so pleased with Dr. Leslie at Cincinnati Children's and we would have never found her if I wouldn't have been connected with others who recommended her so highly. I feel confident that our daughter is getting the best possible care and I know that with the contacts that I have made through the Galactosemia Foundation, I will always have the support to get me through any hurdles I may face. I am so excited to be a part of the Outreach Team and to help connect people to one another!

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*If you are interested in volunteering your time and contact information to assist new galactosemic families – or if you are looking for contacts to learn and share with, contact the Outreach Team at meekfamily1@gmail.com.
Remember to look for our discussion board on Facebook: "Galactosemia Outreach Team Discussion Group

Calling ALL Generation Gs & G Forcers

Are you getting psyched?

Are you saving your pennies, nickels, and dimes?

Are you getting ready for July 2014—the Galactosemia Foundation Conference in Orlando?

If not, now's the time to start thinking about it!

We are busy planning and getting things ready for your big trip and fun times once again with your groups in Orlando!

The GG group and the G Force group will each be split into two groups at the 2014 conference based on age. Even though there will be split groups; they will still do many things together as well.

In addition to enjoying the great things that Orlando has to offer, we will also be doing a bunch of stuff at the hotel—giving each group more opportunities to socialize, share their thoughts and feelings, and enjoy being together, hanging out with old friends, and making new ones.

If you have never been a part of the GG or G Force programs—this is the time! Everyone who has participated in these programs in the past will tell you that it will be one of the best times that you will ever have!

It's often difficult for kids with galactosemia to go into new situations where they don't know anyone—but not to worry—even the most shy people who join in the programs make lifetime friends! This is not like anything else you will ever do because EVERYONE in your groups will have galactosemia, just like you. Only you can understand what it's like and now you will have the opportunity to meet others who also understand.

For those of you who have been part of GG and G Force in the past, this is the time to reach out on Facebook, e-mail, or text and try and get everyone who's your age to come to Orlando and join in making memories of a lifetime!

More updates will come in future newsletters but in the meantime, get psyched, start saving, and get ready for the best time ever in Orlando in July of 2014!!!

We are also open to suggestions, if there's something in particular you would like to do, please let us know! (E-mail, call, or text!)

Generation G (GG)—Contact Linda Manis: Lmscript1@aol.com or 954-610-3739

G Force—Contact Jeannine Quam: Jeannine_Quam@edenpr.k12.mn.us
or 612-232-0171

Calling ALL kids between the ages of 9 and 12 The Mighty Gs!!!

At the last conference we initiated a new program for kids from 9—12 years of age. We would like to see this program expand out of child care and into their own group with two coordinators. If you are a professional and/or parent who would like to coordinate this program, please contact Michelle Fowler.

We have seen the tremendous support that the GG and G Force programs have provided to kids with Galactosemia, and realize that it's time to offer the same opportunities to kids who are younger.

The Mighty Gs will participate in many of the on-site activities as the GGs and G Forcers like they did at the last conference—but this time we hope to offer even more.

So, if you have a Mighty G, we are open to your ideas and suggestions!

We also hope to offer more galactosemic-specific activities to those kids who are under the age of 9—once again, help and suggestions are needed.

We hope to hear from you!

AGERS

During the last conference in July 2012 adult galactosemics had the chance to get to know other adult galactosemics. This was a great time and we enjoyed it. We got to share our own experiences and tips with each other.

During the next conference in July 2014, we will have another chance to get together in Orlando, Florida. The conferences now have something for everyone. This will be another fun and great event for the adults. We welcome all adult galactosemics to join us, since this is a great way to support each other.

As adults with galactosemia, we share our own connections and feel like family. During the conference, we will also feature adult guest speakers. Feel free to join us and share commonalities and similar experiences. In addition to sharing your own experiences you will have a chance to learn from other adults with galactosemia. We hope all adults will join the AGERS during the conference in 2014! Don't miss your chance to sign up for this group! We look forward to a great conference and seeing you there. This conference will bring us together as a group. Contact agers@galactosemia.org via email for more information!

Raising Happy Children

Regardless of a galactosemia diagnosis or not, as parents, we all have the same goals for our children. We want them to be happy, to love/ be loved, follow their dreams and ultimately find success in life. Although we offer our children the same amount of love, opportunities and encouragement at home, siblings very often have different temperaments. Although temperaments are genetic, research shows that happy children are the product of a happy and optimistic home environment.

Here are simple strategies that will help our children thrive and find happiness:

~All children need to receive unconditional love from an adult. Another form of love can be the feeling of being "connected". We need to help our children feel connected -- to us, our family members, friends, neighbors, even daycare providers.

~Our moods matter! Children absorb everything from us. We are modeling behaviors they will replicate later in life. One of the best things we can do for our child's emotional well-being is attend to ours. Children will follow our lead to carve out time for rest, relaxation, and problem solving.

~Children who achieve the feeling of mastery through practice and discipline will have self-esteem, confidence, initiative and an enduring desire to work and accomplish a task.

~The feeling of being valued by others enhances a child's sense of belonging and of contributing.

~We can't prevent stress in our child's life, but we can help them learn healthy ways of dealing with it.

~Our children don't need more "stuff." Over-indulged children are more likely to grow into teenagers who are bored, cynical, and joyless. We need to teach, allow and promote the joy of free play; let's teach our children to entertain themselves by using their imagination. When our children learn to "play in their own mind," they learn to daydream. This is a crucial talent that most children have. We use dreams to chart the courses we take in our lives.

~Help others. Research shows that people who have meaning in their lives feel happier, than those whose lives do not have meaning. Helping others can help make life more meaningful. This can come in the form of helping collect school supplies for children who lost their homes in a natural disaster, helping out with simple household chores, helping an elderly neighbor, or donating clothing to less fortunate people.

Fundraising News - Grant's Wish

The Wilburn Family of Wilmington, IL sponsored Grant's Wish 5k for the 4 th consecutive year on May 4, 2013. The Wilburn's 6 year old son, Grant, has classical galactosemia and they are passionate about furthering research and education in the field of galactosemia. The Wilburn's are very involved in the regional group GAMA. GAMA and the Wilburns raised OVER $8700 at this event. If you would like any information about how you can do the same, please e-mail acredwilburn@att.net!

Living With Galactosemia

Gerard T. Berry, MD and Stephanie Newton, MS, CGC

For many years physicians were suspicious that older adolescents and adults with classic galactosemia, especially men with GALT deficiency, were not enjoying the same quality of life as unaffected individuals. The 2004 paper of Dr. Annet Bosch et al from the Academic Medical Center University of Amsterdam and the 2004 paper of Lambert and Boneh from the University of Melbourne were the first to discuss the consequences of living with galactosemia. They concluded that having galactosemia negatively influences the quality of life. Later, the Dutch investigators reported that the quality of life for patients with galactosemia was not comparable to that in patients with phenylketonuria, another chronic metabolic disease that requires lifelong diet therapy. Subsequently, in our own study of adults with galactosemia, we, for the first time, recognized that 67% and 39% of the adults suffered with anxiety and depression over their lifetime, respectively. This was an "eye-opener" of sorts. What are the implications of this? Several possibilities come to mind. The first is that if half or more of patients with classic disease are anxious and depressed, how likely is it that they would be able to experience rewarding social interactions and meaningful relationships? The second is that a chronic loss of self-esteem may lead to impairment in social interactions, failed relationships and, ultimately, anxiety and depression.

There is a great need to answer these questions. To that end, Stephanie Newton, MS, CGC, is mentoring a current genetic counseling student to conduct a new study. The aim of the study is to begin to understand how patients with galactosemia form and maintain relationships. This new study is needed because past research has not examined if and how anxiety related to speech and language difficulties affects the development of social relationships. This new study will aim to better understand how men and women with galactosemia form meaningful connections in their adult lives. The study will be conducted as a survey. Any individual with galactosemia, over the age of 18, will be welcome to participate. Further details regarding how to sign up to participate will be distributed through the Galactosemia Foundation in the fall, so keep your eyes peeled!

References and resources:

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6. Waisbren SE, Potter NL, Gordon CM, Green RC, Greenstein P, Gubbels CS, Rubio-Gozalbo E, Schomer D, Welt C, Anastasoaie V, D'Anna K, Gentile J, Guo CY, Hecht L, Jackson R, Jansma BM, Li Y, Lip V, Miller DT, Murray M, Power L, Quinn N, Rohr F, Shen Y, Skinder-Meredith A, Timmers I, Tunick R, Wessel A, Wu BL, Levy H, Elsas L, Berry GT. The adult galactosemic phenotype. J Inherit Metab Dis. 2012 Mar;35(2):279-86.

Modifier Genes and Epigenetic Effects In Galactosemia

Gerard T. Berry, MD

Hereditary galactosemia is due to a deficiency of the enzyme, galactose-1-phosphate uridyltransferase (GALT), which catalyzes the conversion of galactose-1-phosphate to uridine diphosphate galactose. Over 230 naturally-occurring mutations in the GALT gene have been described and there are at least 3 major phenotypes: classic galactosemia, clinical variant galactosemia, and the apparently benign Duarte or biochemical variant form of galactosemia. Only the classic form with absent or barely detectable GALT activity is associated with a potentially lethal multi-organ disease state with E. coli sepsis in the newborn period and chronic long term diet-independent complications. Newborn screening has largely eliminated the acute lethal neonatal component, but chronic complications involving the brain and, in females, the ovary, occur even in patients treated prospectively on day one of life. The cause and treatment of these complications is now the single biggest issue in this field. None of the well-known variables such as the time treatment was started or longitudinal galactose-1-phosphate levels can predict who will suffer these complications. Given the inexplicable variation in phenotype, we hypothesize that modifier genes and/or epigenetic effects play a major role in the phenotypic expression of brain disease in classic galactosemia. We hypothesize that the discovery of modifier genes and epigenetic factors will improve our understanding of disease mechanisms and counseling of the patients and their families, and may identify new targets for improved intervention. Our aims are to identify genes that modify outcome severity in classic galactosemia using an unconventional genetic approach, in which we study neurons in culture (derived from patients' induced pluripotent stem cells) to generate transcriptome and epigenetic data in order to eliminate non-informative genomic DNA mutations/polymorphisms obtained from whole genome sequencing.

The purpose of this project is to better understand the cause(s) of long-term CNS complications that occur in individuals with galactosemia. By achieving this goal, better counseling about long-term prognosis can be provided to families whose newborn infant has been diagnosed with galactosemia. We anticipate this information may also lead to a better treatment of this rare orphan disease.

Toward Improved Long-Term Outcome In Classic Galactosemia

Dr. Fridovich-Keil

Despite neonatal or even pre-natal diagnosis and rigorous life-long dietary restriction of galactose many children and adults with classic galactosemia experience a constellation of significant long-term complications; current intervention is inadequate to prevent these problems. Here we propose a new approach to intervention that follows from data we published recently using a fruit fly model for classic galactosemia, and also from preliminary studies described here using patient fibroblasts and blood samples. The strength of this new approach derives both from the data on which it is based, and also from the idea that it shortcuts the drug discovery process by proposing to "re-purpose" existing drugs or nutritional supplements that are already used safely in humans.

The first Aim described here proposes to identify genes and biological pathways that are expressed differently in patients with mild vs. severe long-term outcomes by comparing gene expression patterns in samples of blood from volunteers in these two groups. In a pilot study of RNAs isolated from four blood samples representing pairs of patients with mild vs. severe long-term outcomes, the biological pathway whose genes were expressed most differently between the patient pairs was oxidative stress response – exactly the same pathway expressed most differently between control and affected animals in our fruit fly model of classic galactosemia. Following replication and extension of this result with larger sample sets we propose to screen existing databases to identify known drugs and nutritional supplements predicted to counter the gene expression or functional changes associated with severe long-term outcomes in patients.

The second Aim described here involves testing the impact of drugs or nutritional supplements predicted from Aim 1 to counter the gene expression or functional changes correlated with severe outcome in patient samples. Considering our preliminary data, the first compounds we will test will be known antioxidants and supplements that counter mitochondrial dysfunction, e.g. vitamin C, creatine, taurine, nicotinamide, and ubiquinone. We will test these and other promising drugs or supplements for impact on biomarkers in patient-derived induced pluripotent stem cells in culture, and for impact on a long-term movement outcome in our fruit fly whole animal model of classic galactosemia. Our goal for these experiments is to lay the groundwork for future studies ultimately bringing the most promising drugs or supplements to clinical trial

Dear Galactosemia Foundation Members and Families:

First, I want to say THANK YOU, THANK YOU, THANK YOU, to all the wonderful volunteers and families who have participated so far in our ongoing research study "**Bases of pathophysiology and modifiers of outcome in galactosemia**." Without you, none of this work would be possible.

Research Update: In the past year we completed and published eight research projects related to galactosemia. Some of these projects addressed aspects of outcome in patients; others derived from our work with a fruit fly model of galactosemia. If you would like to see any of these papers or discuss our new findings please let me know and I will be happy to help you get copies of the papers and/or speak with you about them.

Next steps: We are also now beginning to explore aspects of gastrointestinal (GI) health in classic galactosemia. Those who are already enrolled in our research study should have been contacted recently and invited to complete an online survey of GI Health.

We are still actively recruiting more volunteers. If you have not yet been contacted or have not yet completed the survey and would like to do so, please type the address below into your web browser. It will take you to a short online consent form and then a short survey. http://survey.emory.edu/fs.aspx?surveyid=2b5135cbaa84851a6b5ca0f354bb22e
If you have any questions or concerns please let me know; my contact information is listed below.

Please understand that we need respondents who have classic galactosemia, and also respondents who do NOT have galactosemia (e.g. unaffected siblings). Every respondent makes the study more meaningful, whether or not they have galactosemia. Of course, every respondent must complete a consent form before they fill out the survey. As above, if you have any questions or concerns please let me know; I will be happy to speak with you.

Thank you so much!

Judith L. Fridovich-Keil, PhD
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Characterization of a New Mammalian Animal Model of Classic Galactosemia

Dr. Lai

In humans, inherited deficiency of galactose-1-phosphate uridyltransferase (GALT) activity can lead to a potentially lethal disease called Classic Galactosemia. If galactose is not withdrawn from the diet of the affected infants in time, they will suffer from a range of acute toxicity syndrome and die. Consequently, all 50 states in the U.S. include this metabolic disorder in their newborn screening programs. Although a galactose restricted diet, which is the current standard of care, can prevent the neonatal lethality of this disorder, many well-treated patients continue to develop debilitating complications like mental retardation, growth restriction, premature ovarian insufficiency (POI), and other neurological deficits.

Despite decades of research, the pathogenic mechanisms of the acute toxicity syndrome and the long-term complications associated with this disorder remain largely unknown, thus further hampering the development of more effective therapies. One major obstacle in delineating the precise pathogenic mechanisms has been the lack of animal models that recapitulate the organ-specific dysfunctions in human patients. Armed with extraordinary insights, Professor Nancy Leslie and colleagues constructed the world's first GalT gene-knockout (KO) mouse model to address this deficiency. Paradoxically, initial, albeit by no means exhaustive, examination of these animals did not reveal any human disease phenotypes.

Recently, the Principal Investigator (PI)'s research aimed to develop a novel therapy for Classic Galactosemia has resulted in the need for a mammalian animal model for on-going studies. Despite the seemingly lack of patient disease phenotypes in the old GalT-knockout (KO) mouse model, the PI rationalized that it remained a model of choice because of its well-defined genetics, its mammalian nature, and the reported accumulation of galactose metabolites in these animals. Moreover, we cannot emphasize enough that the characterization of the old mouse model was never meant to be exhaustive, and it is too early to abandon the model after a couple of years of studies. Last but not the least; even if the KO animals did demonstrate some degrees of resistance to galactose toxicity, it does not automatically make them worthless research tools. On the contrary, if we can decipher the mechanisms utilized by these mice to resist/ minimize galactose insults, we could identify the molecular targets of galactose toxicity in human patients, and to design better therapeutic options.

When the PI contacted Professor Leslie and requested the sharing of the old GalT-KO mouse model, he was saddened to learn that the model no longer existed. However, this did not deter the PI's resolve to advance. In response, the PI used a similar technology to construct a new GalT-KO mouse model. Although these new GalT-KO mice, like the old GalT-KO mice, manifested some degrees of resistance to galactose toxicity, subtle phenotypic differences between the KO mice and their wild-type (i.e., normal) littermates do exist. In this application, we propose new studies to characterize these subtle differences at the molecular and biochemical levels, as well as to understand the basis of galactose resistance in these animals.

Nutrition Management of Classical Galactosemia – Further Food Analysis and Meeting Educational Needs

Dr. VanCalcar

This project will continue activities initiated with a Galactosemia Foundation grant awarded in 2011. Additional analysis of galactose in remaining questionable ingredients and foods will be completed. These include legumes, tofu, fermented products made from soy and some additional cheeses including mild and medium cheddar cheese. Also, new recommendations from the Galactosemia Task Force, which was formed to evaluate current research about diet for classical galactosemia, will soon be published in a medical journal. Since diet recommendations have changed, information about these changes needs to be provided to professionals, families, and individuals with galactosemia. Thus, new educational materials will be developed for metabolic clinics to use with new families, parents, and children, teens, and adults with galactosemia.

Before E-mail & Before Internet
Parents of Galactosemic Children (now the Galactosemic Foundation) Was Formed

by Linda Mannis

Many of you who attended the conference last year in Dallas probably didn't realize that three of the four original PGC Board Members were in attendance. Linda Manis, Christine Kovach, and Rhonda Bauer—our only missing link was Gayle Dennis.

I was so frustrated 29 years ago with only being given 1 sheet of paper about galactosemia from our metabolic clinic in New York City! One of the nurses at the Clinic agreed to address (they couldn't give addresses to me) and send out stamped letters I gave her inviting families for lunch at my house in New York. And that was the beginning.

Over a 3-year period, Parents of Galactosemia took the next step and became an official nonprofit organization. Below is an excerpt from our February 1989 Newsletter (Vol. 4, No. 1) along with the photo that accompanied the article.

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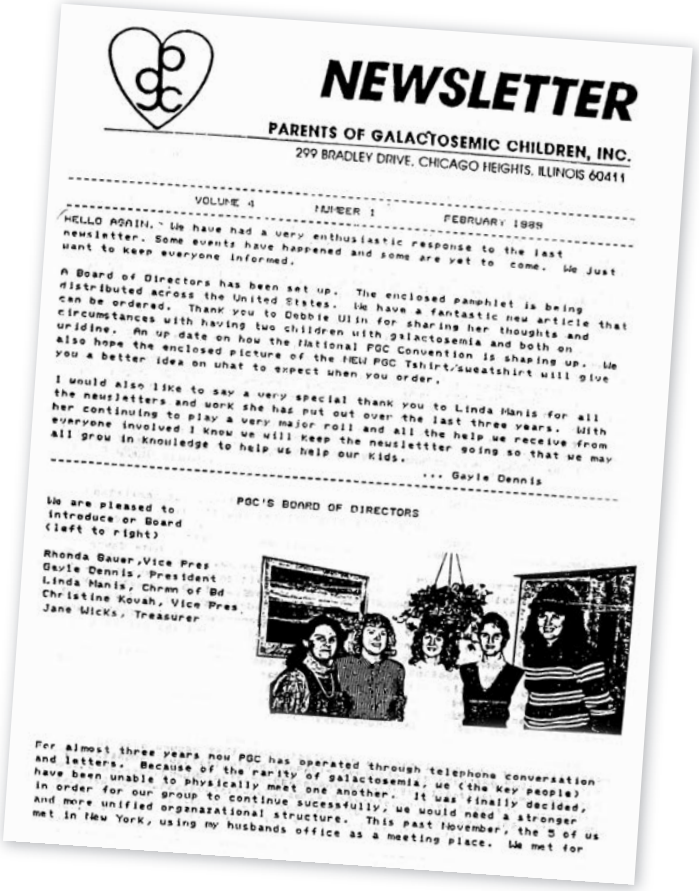
For almost 3 years now, PGC has operated through telephone conversations and letters. Because of the rarity of galactosemia, we (the Key people) have been unable to physically meet one another. It was finally decided, in order for our group to continue successfully, we would need a stronger and more unified organizational structure. This past November (1988), the five us met in New York, using my husband's office as a meeting place. We met for two wonderful days. It was great meeting one another for the first time and sharing our experiences and information.

We wrote the first PGC pamphlet and five copies were sent to each Metabolic Clinic in the country in January (1989). The Board consists of:

Linda Manis, Chairman of the Board (New York)
Gayle Dennis, President (Ohio)
Christine Kovach, Vice President (Oregon)
Rhonda Bauer, Vice President (Texas)
Jane Wicks, Treasurer (New York)—Jane left the Board shortly after this.

There are a few other people who have put a lot of work into PGC that should also be mentioned: Barbara Sprow, Mary Rosner, Nate Bauer, Karen Mitchell, Debbie Ulin, Diane Gomes, and Sally Ferry

Photo (Left to Right: Rhonda Bauer, Gayle Dennis, Linda Manis, Christine Kovach, and Jane Wicks):



NEWSLETTER

PARENTS OF GALACTOSEMIC CHILDREN, INC.
299 BRADLEY DRIVE, CHICAGO HEIGHTS, ILLINOIS 60411

VOLUME 4 NUMBER 1 FEBRUARY 1989

HELLO AGAIN. We have had a very enthusiastic response to the last newsletter. Some events have happened and some are yet to come. We just want to keep everyone informed.

A Board of Directors has been set up. The enclosed pamphlet is being distributed across the United States. We have a fantastic new article that can be ordered. Thank you to Debbie Ulin for sharing her thoughts and circumstances with having two children with galactosemia and both on uridine. An up date on how the National PGC Convention is shaping up. We also hope the enclosed picture of the NEW PGC Tshirt/sweatshirt will give you a better idea on what to expect when you order.

I would also like to say a very special thank you to Linda Manis for all the newsletters and work she has put out over the last three years. With her continuing to play a very major roll and all the help we receive from everyone involved I know we will keep the newslettter going so that we may all grow in knowledge to help us help our Kids.

... Gayle Dennis

PGC'S BOARD OF DIRECTORS

We are pleased to introduce or Board (left to right)

Rhonda Bauer, Vice Pres
Gayle Dennis, President
Linda Manis, Chrm of Bd
Christine Kovah, Vice Pres.
Jane Wicks, Treasurer

For almost three years now PGC has operated through telephone conversation and letters. Because of the rarity of galactosemia, we (the Key people) have been unable to physically meet one another. It was finally decided, in order for our group to continue sucessfully, we would need a stronger and more unified organazational structure. This past November, the 5 of us met in New York, using my husbands office as a meeting place. We met for

Life changes and so did the Board, our kids are now grown, but for those of us in the beginning, we have fond memories of one another and those hopeful days in the beginning that we would create a respected organization not only by families but by the medical community.

We are so proud of our "baby"—because of the many countless hours spent by volunteers who have followed in our footsteps, the organization has become a worldwide, well-respected organization.

And although, we have lost touch with some of the early volunteers, it is amazing when we do reconnect at the conferences and see how far this organization, started by a few moms who had a dream has come.

2013

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

APRIL	MAY	JUNE
	17 Fore The Cause Golf Outing, Richmond, VA	7 & 8 GFSS Social, Decatur, TX 9 Kyleigh's Cure Road Race, Richmond, VA 15 Race For Jase, Crosslin Park at 8 AM, Enid, OK

JULY	AUGUST	SEPTEMBER

OCTOBER	NOVEMBER	DECEMBER

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www.galactosemia.org

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