



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

In Memoriam

Professor Louis Jacob Elsas II (10th Feb., 1937 – 16th Sep., 2012)



(Photo: Courtesy of
University of Miami
Miller School of
Medicine)

Professor Louis Jacob Elsas II, a beacon for the metabolic and genetic community died on September 16th, 2012. Dr. Elsas or Skip, as he was known to all of his friends, was born on 10th February, 1937 in Atlanta, Georgia to Mr. Herbert Rothschild Elsas and Mrs. Edith Levy Elsas. He attended Phillips Andover Academy, before he entered Harvard College and earned his B.S. in Biochemistry in 1958. Upon graduating with an M.D. from the University of Virginia in 1962, Dr. Elsas set his eyes at the academic world where he excelled as an educator, a scientist and a physician. After his post-graduate training and an early faculty career at Yale University, Dr. Elsas' never-ending desire to return to his family roots led to his acceptance of a faculty position at the Emory

University School of Medicine in 1970. Armed with extraordinary intellectual talents and energy, Dr. Elsas quickly established the Division of Medical Genetics in the Department of Pediatrics at Emory and initiated the first state-wide newborn screening program in Georgia. Under Dr. Elsas' leadership, the Division of Medical Genetics at Emory became one of the premier programs in the United States. With his pioneer initiative in newborn screening, countless lives inflicted with metabolic disorders such as Classic Galactosemia were saved in the decades that ensued. Dr. Elsas also had a longstanding research interest in metabolic disorders, including Classic Galactosemia. He authored more than 250 scientific papers and received countless research grants, honors and awards for his scientific accomplishments. Last but not the least Dr. Elsas was the Founding President of the Society for Inherited Metabolic Disorders (SIMD), which had just held its 34th Annual Meeting earlier this year.

In 2002, Dr. Elsas retired from Emory at the age of 65 and launched his second career to build another world-class genetic program by becoming the first Director of the Dr. John T. MacDonald Foundation Center for Medical Genetics at the University of Miami Miller School of Medicine. In 2012 when his cancer deemed overwhelming, Dr. Elsas finally stepped down and moved back to Atlanta with his wife. A loving, dedicated husband, devoted father, and proud grandfather, Dr. Elsas is survived by his wife Nancy Terrell Elsas; his three children and five grandchildren.

He will be missed by all, especially those in the galactosemia community.

A Louis "Skip" Elsas memorial fund has been set up with the Galactosemia Foundation.

This edition of the newsletter
is dedicated to Louis "SKIP" Elsas
by the Galactosemia Foundation

Parents of Galactosemic Children, Inc. is a non-profit charitable organization. Founded in February 1985 by a small group of mothers in New York, PGC realizes the need for further information and networking between affected families and professionals. Metabolic Clinics across the nation continue to assist PGC 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.

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PGC 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

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A Note From Your President

Galactosemia Foundation Board of Directors and 2012 Conference Committee members are just beginning to relax from the high energy of our 2012 Texas Conference. A huge thank you is extended to each and every person that attended this event; from families to friends to professionals to vendors, each and every person made a difference and much appreciation is expressed; it takes everyone working together to have such a successful event as we experienced. GFSS was our host group for this event and they did such a wonderful job, thank you!

We have been busy with continuing to move forward in many ways. As you will see detailed in this newsletter, we are currently engaged in accepting research grant proposals for our next round of research funding. We are working closely with professionals in an effort to help them gain additional funding for grants and proposals through our support. Many fundraising efforts are in preparation by several families to help boost the funding in our accounts so we can continue to grow and help families living with galactosemia. Our 2014 Conference Incentive program is already underway as well.

As we all know, social media has changed in so many ways over the past few years. Galactosemia Foundation continues to integrate emerging social media technologies into our plan in an effort to reach as many of the health and genetics communities, friends and families and professionals as possible, through Facebook and our continually updating website. We are also active participants in both Goodsearch and Goodshop which are both excellent, easy, free ways to help support the foundation; especially this holiday season. So please, take a few minutes of your day and join our Facebook, visit our website from time to time and make Goodsearch and Goodshop a part of your everyday internet searches!

Wishing everyone a safe, joyous holiday season as Galactosemia Foundation enters 2013 reflective about what many years of innovation have given and what we take away to move forward with. We head into the future excited about many projects and working with each and every one of YOU! We are striving to focus on the broad network of our galactosemic family.

Blessings to all,
Michelle Fowler
Galactosemia Foundation President

New Board Members

New board member, Theresa Boehm, is a committed mother of three children and one grandson. She has one child with classic galactosemia, 21 year old Blake Boehm. Theresa brings to the foundation knowledge in regards to non-for-profit organizations, grant writing, financial background, project coordinating, convention coordinating, as well as a great work ethic. Theresa looks forward to working with the other board members to make the foundation as successful as possible.

Scott R. Saylor is extremely pleased to join the board to serve The Galactosemia Foundation. From the time their son Jake was diagnosed with Classical Galactosemia in 2007 the Saylor family has set out to raise money and raise awareness of Galactosemia. Scott earned a B.S. in Management and minors in Human Resource Management and Marketing from Gannon University, Erie PA where he was also a 4 year starter in football. Scott works as a District Manager for Walgreen Co. in Central Virginia. He is responsible for the financial and operational results of 41 stores and 300 million in annual sales. He has been with Walgreen Co. for 17 years. Scott resides in Chesterfield, Virginia with his wife Kristine and their children Brooke(8) and Jake(5). Jake Saylor was born in 2007 and was diagnosed 10 days later with Classical Galactosemia. Scott and Kristine are both originally from Ohio and enjoy rooting for all Ohio sports teams along with rooting on their children in many different sports.

Treasury Report

Galactosemia Foundation's 2012 "Empower Our Future"

Texas Conference - Treasury Report

INCOME:

| | |
|-----------------------------|-------------------|
| Donations | \$8,270.32 |
| Adult Registration | \$21,683.54 |
| Child Registration | \$5,461.50 |
| Fundraising Incentive | \$88,035.45 |
| G-Force | \$6,209.14 |
| Generation "G" Registration | \$4,256.19 |
| Sales & Raffle | \$10,566.96 |
| <u>Sponsors</u> | <u>\$6,000.00</u> |

Total Income: **\$150,483.10**

EXPENSES:

| | |
|--|--------------------|
| A/V | \$19,484.48 |
| Childcare | \$14,810.00 |
| Conference Hotel | \$70,796.20 |
| Conference Planning Meeting | \$10,660.23 |
| Friday Night Social | \$1,663.99 |
| Fundraising Incentive | \$5,080.17 |
| Generation "G" & G-Force Program | \$11,311.46 |
| Insurance | \$393.00 |
| Office Supplies | \$924.03 |
| Postage | \$1,249.72 |
| Printing | \$3,971.19 |
| Registration | \$3,613.98 |
| Sales Items | \$7,289.75 |
| Saturday Night Social | \$3,282.83 |
| Signs & Banners | \$1,516.19 |
| <u>Travel/Lodging (Board & Speakers)</u> | <u>\$22,532.25</u> |

Total Expenses: **\$178,579.47**

OVERALL TOTAL: **\$(-28,096.37)**

As you can see after you factor in the expenses and income the conference cost GF \$28,096.37. Thanks once again to all that participated in the conference incentive program. Because of this the conference costs to the organization were only slightly higher than the 10' conference which cost GF \$21,573.16. Our 2014 conference incentive program is now open! This is a great way to raise funds for the 2014 Orlando conference and can help you offset some of your travel expenses. Please keep in mind that in order for your donations/fundraising to be considered for the incentive program they must be marked and deposited into the General account. If you have any questions on the incentive program please email me.

GF's account balances as of 10/24/12 are General Account \$11,248.26 and Research Account \$136,078.31. In order for GF to continue to hold conferences donators need to remember the General account. Help us reduce costs by registering with our new on-line database which will allow families to update addresses, phone numbers, and email plus you also have the option of receiving future newsletters via email. Newsletters are a huge expense to print and mail. If you would like a complete breakdown of the report please send a written request to me at treasurer@galactosemia.org.

Paul Fowler

The Generation G Page

(GGs: The 1st Generation of Galactosemics)



More than 30 young adults participated in the GG program at the Dallas conference. We kicked off the program with lots of laughs with Comedian/Magician Doc "Bo" Holiday, who chose volunteers from the audience to assist in all kinds of funny antics.

The No Whey Café—Texas style—was used by both the GG and G Force programs.

Role-Playing Menu Reading: Switching off as servers (complete with aprons and guest checks) and customers who ordered from real restaurant menus. The idea of course was for GGs to experience a server's perspective with food ingredient inquiries while providing GG customers an chance to practice ordering dairy-free meals.



Cooking & Tasting Demo—Intern nutritionist "Rachel" (who works with Sandy VanCalcar) wins the award for the Top No Whey Café Chef. Along with volunteers, Rachel demonstrated how to make 5-Minute Dairy-Free Fudge and Tex-Mex Snack Mix—everyone got a "taste." Then, stations were set up for groups of four and

using sandwich makers, they made their own Grilled Chocolate & Banana sandwiches.

Galactosemia Jeopardy—Who knew how much everyone knows about galactosemia—I don't think there was one question that the GGs could not answer!! Too bad it was not for "real" money!

Sessions with Psychologists & Lunch with the Doctors—There were two private sessions with two different psychologists, who talked with the GGs, giving them the opportunity to share their feelings about what it's like to have galactosemia, how it affects their self-esteem, along with the different problems they have faced during their lives. Both psychologists provided positive solutions and ways in which to deal with various issues. The GGs found these sessions to be the most helpful—we plan to offer even more of these types of sessions at future conferences. At the GG "Lunch with the Doctors" they learned what the latest research is in a variety of fields and were able to ask the professionals questions—our amazing team of conference speakers went out of their way to provide these young people with information about galactosemia, and we thank you.

Offsite Trips—The annual bowling trip as always was a great way for old and new GGs to get reacquainted. Stockyards Station on the second day was a great hit once we got to the mechanical bulls (almost everyone had a ride)—the photos tell it all.

Then, the Wild West Bill Show—gave us a chance to cool off (it was 107 outside)—and experience the old West.

The Dance on Saturday night capped off a great 2½ days of fun—the lifetime connections and friendships that were made could be seen as the GGs danced the night away together!

We had a number of GGs who had not been to a conference before—it's always so amazing to see these shy young guys and gals at registration—who don't know anyone—and as the hours and days go by, they are making new friends, talking and getting to know one another. The courage it takes for GGs and G Forcers to participate in a program where they do not know anyone speaks volumes about just how important it is to them to meet others their age who also have galactosemia.

This was the fifth GG program (4 at conferences and 1 for the adult research study in Boston). And as this group has evolved we see that there's a need to split the GG group into two groups by age. So basically, it will still be the GG program—but with two groups, who will do things together and separately. In addition, at the GG wrap up, the GGs made a number of suggestions, including having the opportunity to talk to the parents of children with galactosemia (in small groups), and also talking to the younger kids and teens with galactosemia. These are among some of their great ideas, which we will incorporate in the future. More details will be provided as we get closer to the Orlando conference.

And finally, I would like to personally thank the Galactosemia Foundation, the Texas group volunteers, and my GG parent volunteers for all their help in making the GG program such a huge success! But mostly, I would like to thank the Dallas GG group for being such a wonderful group of young people! I feel so lucky to have gotten to know each and every one of you—thank you!

For more information on the GG program, please contact:

Linda Manis (Lmscript1@aol.com)

AGERS Adult Group

by Kimberley Malyn

Starting in 2012 there was a new group at the Galactosemia Foundation Conference. It was the Adult Galactosemic group. We had a small group of about twelve adult galactosemics and some of us brought our spouses.

We all had a chance to get to know each other. Everyone got to tell a little about themselves. Some of us also talked about our own experiences with being diagnosed, eating in restaurants, and having families.

We all felt like we could ask questions about anything from diet to additional health issues to getting and keeping jobs, etc. We all felt like we could contribute to answering questions because we all have our own experiences.

Some of the group participated in the research projects during the conference. Some of the group also went to dinner together one night.

We also feel like it is important to talk with other people at the conferences such as parents of galactosemics that have questions about our experiences, etc. so we also got to spend some time at the regular session and breakout sessions.

I like to bring my own children to the conferences because I know it encourages other galactosemics to have families and I tell people about my family. My children also enjoy the conferences. They enjoyed the activities a lot. They got to meet children their own age and they also had a lot of fun at the evening family activities.

At the next conference I hope this group is even more diverse. I hope to see even more adults at the next conference to meet and share stories. I always feel encouraged and enjoy encouraging other people when I attend the conferences. Since galactosemia is rare it is hard to meet other galactosemics. I remember how great it felt when I attended my first conference because up until then I had thoughts such as "Am I the only person in the world with galactosemia?" It feels great to be able to go to the conferences and now we have groups that will help us get to know each other better and help us stay in contact with each other. I would encourage you to come to the conferences. I want to thank the Galactosemia Foundation for forming new groups and trying to help everyone get with groups of their own ages so that everyone can meet others close to their own age.

G-Force Texas Style

Howdy Partners! Almost 50 G-Forcers came on down to Dallas, Texas to "Join the Force" for some southern fun! It was our second launch as teens from around the world enjoyed various activities with Texas flair.

Our galactosemia conference began with a lot of laughs, as we watched comedian, Doc Holiday present his magic. We got a kick out of seeing our friends up on stage in the act! It was meaningful to make new friends, while getting reacquainted with our old pals.

Both on and off site activities were enjoyed by all. We had the chance to talk with psychologists who have family members living with galactosemia themselves. G-Forcers participated in activities which were designed to promote self-esteem, a sense of friendship and support. We also learned new information while playing, "Galactosemia Jeopardy". A true sense of excitement filled the room, as we played boys against the girls!

Having galactosemia presents food challenges, thus teens were given the opportunity to role play at the "No Whey Café". Everyone had a fun time being a server or customer to learn how to ask good questions while dining out. Teens also enjoyed cooking and of course tasting the dairy-free fudge, a Tex-Mex snack mix and grilled chocolate banana sandwiches!

It's always an adventure taking a bus to a new place especially when the temperature is 107! Our trip to Stockyards Station, Rodeo Zone and The Wild West Bill Show gave us a good feel for Texas. Of course we



cannot forget the fun we had bowling with our friends at The Main Event and dancing the night away with friends with whom we are "Linked for Life". We look back at our Texas G-Force launch with fond memories and look forward with great anticipation to our next national galactosemia conference in Orlando, Florida!

One Child's Story

by Michelle Fowler



Many parents ask, "What is your child's story and what do you do for your child?" We would like to share a little bit about what we do with our son both as information and encouragement!

Landon Fowler, son of Paul and Michelle Fowler from Mandeville, Louisiana is classic galactosemic. Landon has an older sister, Tori (??) and a younger sister, Tela (carrier). Landon celebrated his 13th birthday on October 13th and enjoyed a homemade chocolate cake with peanut butter icing; of course, galactosemic-safe! Having attended galactosemic conferences, Landon has made many galactosemic friends and has also gained the desire to encourage families with galactosemic children in a positive manner. Landon says if he had the choice, he would not want to change being galactosemic, because that is what makes him who he is.

His parents were told he would never play sports because of possibly having brittle bones due to galactosemia. Landon proudly represents his junior high football team. Landon plays both offensive tackle and defensive tackle for the 7th grade team.

His parents were told he would be left out of many events at school where eating was involved. The football team eats before each game where they are served chicken pasta, of course, not galactosemic safe. His parents bring Galactosemic safe, chicken pasta for Landon to enjoy and most of the boys never even notice his is different!

His parents were told he may feel left out from not eating lunch at school. Landon is not allowed to have anything other than water from the school and he does take his lunch everyday to school. However, Landon says many of his friends bring their lunch too and he would rather take his lunch to school than eat in the cafeteria.

His parents were told not to allow him to participate in Halloween since he would receive mostly candy he could not have. Halloween is spent trading the un-safe candy for safe candy with his sisters and receiving quarters from his parents by selling the candy he cannot have! This money is donated to the Galactosemic Foundation.

His parents were told he would not be able to enjoy many foods and would be a picky eater. Well, Landon's favorite food is lasagna; this is made with noodles, meat sauce with a gravy base and topped with Dayia cheese. He has a substitution for almost everything!

His parents were never told anything about his teeth. Well, Landon has trouble with the enamel on his teeth. His braces had to be applied with special glue due to the 'usual' glue not sticking to his teeth. This also holds true with cavities and sealant, things do not 'stick' to the teeth as they should.

His parents were told he would require special education in school as well as speech therapy. Well, Landon is involved in talented art and maintains the honor roll with his grades. Landon is an ace at many X-box games, loves playing outside and spending time with his family and friends.

Through the years, rather than focusing on what he can't do; Landon focuses on being a kid, just like every other thirteen-year-old!

Who do you know?

Dr. Fridovich-Keil remains active in seeking funding for galactosemia research. The US Department of Defense has research funding opportunities available for various topics. Many of the subjects which receive funding are congressionally mandated, meaning that a member of Congress has a special interest in that topic, allowing it to be put on the funding list. The website is: <http://cdmrp.army.mil/funding/default.shtml>.

If a member of Galactosemia Foundation has any congressional ties, it would be beneficial if galactosemia, metabolic disease, or even disorders detected by newborn screening could find their way to this list. Obtaining additional funding for galactosemia research is going to take work as the NIH budget continues to face cuts. Making connections and keeping galactosemia in the forefront for research dollars is vital.

Self Esteem

by Andrea Topper

Positive self-esteem is crucial to our children's development. It is the foundation of everything they do, everything they are - it is the foundation of their future...and OUR future! It is an important ingredient for a happy life. We are the primary influence on how our children feel about his/herself and their self-esteem. Who they are is a mirror of US!

As parents, we cannot control everything our child sees, hears or thinks which will be contributing to his or her self-image. But there is still much that we could do. As caregivers, we could listen, accept, discipline, and set limits for good self-esteem. Allow independent decisions and offer acceptance in order to develop positive esteem.

Self-esteem is built by realistic praise. Our children know when praise isn't real. It is important, that compliments and praise be credible. Exaggerated compliments like, "You are the best kid in the universe. You are the nicest person that ever lived" can actually be counter-productive. The child will develop an inflated ego, and that can affect his relationship with friends, which in the long run will have a negative effect on his or her self-esteem.

An important contributing factor to a child developing self-esteem is an adult who helps the child feel special and appreciated; not ignoring a child's problems, but rather focusing on a child's strengths.

It's important for us to set aside "special alone times" each week with each of our children. It's a good idea to tell our children "When I read to you, talk to you, or play with you, I won't even answer the phone if it rings." During these special times, focus on things that our children enjoys doing, giving them the chance to relax and display their strengths. On a regular basis, we must remember to tell

our children how fortunate and how proud we are to be their parents. We should talk positively about our children in the presence of important people in his life, such as grandparents, teachers, friends etc.

High self-esteem brings solid problem-solving skills. We need to help our children reflect upon possible solutions. Try role-playing situations with our children to help demonstrate the steps involved in problem solving. Discuss ways for them to explain Galactosemia to their peers, teachers or a waiter/waitress at a restaurant.

We have to give our children choices. This lessens power struggles. Ask our children if they would like to be reminded five or ten minutes before bedtime to get ready for bed. Give them choices regarding safe foods they can enjoy. Allow our children make dinner selections, or help grocery shop. These choices help to set the foundation for a feeling of control over their life.

Remember to highlight your child's strengths. Most kids see themselves negatively - especially at school. List your child's areas of strength. Select strength - reinforce and display it. At the beginning of the school year, speak with your child's teachers and tell them what your child's special strengths are and about the areas in which he or she excels, so the teacher will have a positive outlook towards them and will continue to build on those strengths. Every child has numerous gifts and it's our job as parents to identify them!

Set realistic expectations and goals for your children to give them a sense of control.

There are many ways we can help build our kids' self-esteem. The first and most important way is by demonstrating our love for them on a constant basis. The more kids feel loved, the better their self-esteem will be.

NIH Funding

The National Institutes of Health (NIH) is a medical research center. The center receives funding from the government for research on rare conditions, like galactosemia. NIH is in danger of losing much needed funds for research grants.

Families can access a form letter by using the following link:
<http://capwiz.com/faseb/utr/2/?a=62032501&ti=24825561&tc=>.

Feel free to adjust the wording to meet your needs. It is important to keep our Congressmen aware of the importance of funding for rare disorders.

Letter from Scott Shepard, Galactosemia Foundation Vice President

Only a little over two short years ago my wife, Choi, my sister Linda, and I were sitting in the audience of our first Galactosemia conference in Minnesota. I could not imagine at that time that I would be writing to you today as the Vice President of the Galactosemia Foundation. I got to meet and talk with some of you in Dallas in July and I look forward to getting to know many more members of our Galactosemia family over the coming years.

I grew up in a town in rural Missouri with my family (I have an older brother and two older sisters). After graduating from college with a degree in Mechanical Engineering, I moved to Topeka, Kansas to work as an engineer at a Hallmark Cards manufacturing plant. After about 5 years at Hallmark, I moved to Texas to take a job at Frito-Lay and have been here for the past 13 years. While at Frito-Lay I have worked on brands that you may be familiar with, such as Lay's Potato Chips, Frito's Corn Chips and lots of other products (many of which do not contain milk – see <http://www.fritolay.com/your-health/us-products-not-containing-milk-ingredients.html> and of course be sure to read those labels).

My wife and I met and got married here in Texas. A little over six years ago, we had our first daughter, Jasmine. We brought Jasmine home from the hospital for a couple of days and then readmitted her, where she was diagnosed with Classical Galactosemia at nine days old. Those first few weeks were a difficult time for us – we did not know if our baby would survive and what her and our future would be like. Fifteen months later, we had our second daughter, Olivia. Olivia does not have Galactosemia.

Why I volunteer for and donate money to the Galactosemia Foundation

I want my daughter to have the best opportunity to live a fulfilling, productive life. I want her to grow up knowing that she is not the only person in the world with Galactosemia and hopefully develop friendships with other people with Galactosemia. I want to learn from other parents that are further along this life journey and to share what I have learned with parents that are just starting out.

We are very thankful to the people who came before our family and built what has become the Galactosemia Foundation, the regional support groups, and raised the money to fund Galactosemia research. It is amazing to see the progress that we have made by reading the early newsletters and talking to families that have been involved for a long time. When Jasmine was first diagnosed, we were eager to find information about how to raise a child with Galactosemia and what to expect. The prognosis from the scientific articles seemed bleak. Being a part of the regional and national Galactosemia groups gave (and continue to give) us hope, knowledge, and strength.

Early Intervention

One of my interests is to share what we have learned and our passion for advocating for our Galactosemic daughter. Knowing

that Galactosemics are more likely than the general population to have difficulties with speech, language, gross and fine motor skills, we began learning about therapies, special education, and being advocates when our daughter was very young.

Not all Galactosemic children will need early intervention or special education. However, I encourage all parents of Galactosemic infants and toddlers to learn about the early childhood programs available in your state or country and have your children evaluated. There is no downside to having your child evaluated. If they do not need any help, you get the peace of mind of knowing that. If they do qualify, then you can take full advantage of any available programs.

We began taking Jasmine to a Developmental Pediatrician when she was four months old. When Jasmine was eighteen months old, the Developmental Pediatrician referred her to be evaluated by the State of Texas Early Childhood Intervention (ECI) program. In the United States, each state has some form of early childhood intervention programs (they may be called something different than “ECI” in other states) as mandated by the Federal Individuals with Disabilities Education Act (IDEA) part C, which covers infants and toddlers (up to age 3 years). The National Early Childhood Technical Assistance Center (NECTAC) maintains a list of IDEA part C coordinators by state at <http://www.nectac.org/contact/ptccoord.asp>.

Jasmine qualified for ECI services (Galactosemics qualify automatically in Texas). Through ECI, Jasmine and our family received two 45 minute sessions with a speech therapist, two 45 minute sessions with a developmental specialist, and one 45 minute session with a nutritionist each month from 19 months until she was three years old. The great things about these services were that they were free and convenient. The ECI providers would either come to our house or come to Jasmine's daycare facility to work with her one-on-one and in peer settings.

Transitioning from Early Intervention to Pre-School

When your child turns three years old, they are no longer eligible for State run IDEA part C programs. Your local school district is now responsible for providing special education services (under IDEA part B) to eligible children. For children that are participating in IDEA part C (early intervention) programs, the early intervention provider will facilitate transition discussions with your school district.

Navigating all of the laws, rules, procedures, and bureaucracy to develop a successful working partnership with your school district can be complicated – particularly because most school districts are unfamiliar with Galactosemia. Learning all of this stuff may at first seem daunting and even insurmountable – just like the Galactosemia diet and reading food labels may have seemed daunting at first. We have found Wrightslaw (<http://wrightslaw.com/>) to be a helpful resource. I would particularly recommend the book “Wrightslaw: From Emotions to Advocacy – The Special Education Survival Guide, 2nd Edition” to those of you with school age children (3 years and up).

Survey Results from the Conference Provide Great Feedback

Thanks to all who responded

After the July 2012 GF conference, a lengthy, survey was sent to all attendees. Questions ranged from the registration process, the food, conference sessions, layout and entertainment. Over 50 people responded to the survey and provided feedback that we will use as 'lessons learned' for the 2014 GF conference.

62% - of the responders were parents of children with Galactosemia

13% - were Relatives of children or adults with Galactosemia

33% - indicated this was their first conference

30% - have attended 2-3 conferences

34% - have attended 4 or more conferences

Highlights from the survey include

- 92% of attendees felt they 'gained a better understanding of Galactosemia'
- 100% responded they were happy with the Childcare, the GG and G-force programs
- 73% stated they were likely to attend the next conference

Critical feedback included:

1. AGERS program must be improved if done again
2. Must be a more positive tone
3. Better breakfast choices and onsite dining
4. Not enough time for everything
5. Complicated information was difficult to understand

Positive feedback included:

1. Many thanks from attendees for all the volunteers that setup and dedicated their time.
2. Affordable
3. Social was excellent
4. Wide variety of topics
5. Auction was well liked

We asked for input on topics for the next conference as well as what we should 'keep the same'. All of this information will be used for 2014 conference planning.

Thank you to all that responded to the survey.

The full survey response link is included on the website if you would like to review.

Galactosemic Families of Minnesota Update

It was a crisp fall day on Saturday, September 22, 2012 as Galactosemic Families of Minnesota (GFMN) had our second "Walk for Galactosemia" around the beautiful Lake Nokomis in Minneapolis, Minnesota. Approximately 90 supporters both young and old walked for the cause. After the walk we enjoyed a delicious Dairy-Free lunch at the Nokomis Community Center.

GFMN works to provide support to people living with galactosemia in the Midwest, as well as our National Galactosemia Foundation. We would like to extend a warm welcome to anyone, who would like to join our group! We meet formally twice a year in Minnesota in the spring and fall to provide support, friendship and educational information to those affected by galactosemia. Please contact Jeannine Quam at jmquam@edenpr.org for further questions.

GAMA News

The Galactosemia Association of Midwest America (GAMA) met in August to share conference information and of course, EAT! Eight GAMA families enjoyed a safe buffet and face painting, as well as good old fashion playing and fellowship at the home of Seth and Nish Rao.

GAMA will meet again in November for a fall get together! If you would like to join us, please contact our social coordinator, Gioia Lauro-Geruso at glauroge@gmail.com.

GAMA continues to update their website with recipes and resources for families. Please visit galactosemiamidwest.com for the latest information re: GAMA's outings, fundraisers and tips!



Let's Get The Word Out....

I'm using my business card to bring more awareness to Galactosemia. Depending on what your business is, maybe you too can put the "Galactosemia Foundation" on the back of your card!

Thank You PRINT PLUS for doing such a great job!

Thank you EVERYONE for the wonderful GALACTOSEMIA FOUNDATION SILENT AUCTION AND RAFFLE!!! We had items donated by Galactosemic groups, companies, and individuals. It would not have been possible without all of these items and the volunteers that worked so hard to get them donated! THANK YOU, THANK YOU, THANK YOU!!! The silent auction and raffle profited over \$4,000!!!!!! That would not have been possible without EVERYONE OF YOU!!!! All of those funds go straight back into the Galactosemia Foundation budget for 2014 Orlando Florida Conference! I hope you all had as much fun as we did with the silent auction. If anyone has any comments or suggestions for the next silent auction, don't hesitate to email me.

---Kellie Wilcox
kelliekee@yahoo.com

Outreach Team – Fall 2012 Update

Welcome new families!

The Outreach Team has had a busy year fielding new emails from families across the world. We have been in contact with parents of new galactosemic babies from Alabama to Iran and beyond. Embracing these new families is an important function of our wonderful organization. We are always looking for local contacts and enthusiastic team members. If you are passionate about reaching out to new families and providing information and guidance, please contact the Outreach Team to volunteer!

The Outreach Team is for more than NEW families! Regardless of your age or how long you've been with us – if you are searching for more contacts, buddies or friendly families in your local area, please contact the Outreach Team for contacts! We just might know of someone anxious to get to know you and your family too! And just as important, if you would like to be a local contact and are interested in sharing your contact information with other interested galactosemics and their families, please let us know!

The number one request we receive: Where can I get the latest diet guide??

The diet guide is available instantly online at this link:

http://www.galactosemiamidwest.com.php5-24.dfw1-2.websitetestlink.com/wp-content/uploads/2012/02/GALAC.GUIDE_.FINAL_.3.pdf

Find us on Facebook! The Outreach Team has a discussion group, look us up: Galactosemia Outreach Team Discussion Group

*Please remember to sign up with The Galactosemia Foundation online, and be sure that you are receiving the newsletter!

Sign up here: http://galactosemia.org/Join_Newsletter.php

We continually hear how thankful new families are to have an organization, such as The Galactosemia Foundation to reach out to when they are first faced with a galactosemic diagnosis. Many of us remember, very clearly, the first time another parent or another galactosemic reached out to us, or answered the questions we needed answered at 10pm on a Sunday night. The Outreach Team is committed to maintaining these relationships and continuing to connect people and families with galactosemia. We are so grateful to our team and national contacts that continually assist in making these connections successful, THANK YOU!

To volunteer please contact Robyn & Jeremy Meek at meekfamily1@gmail.com

Galactosemia Foundation Research Team Update

The GF Board of Directors has approved a 4th round of research grants totaling \$75,000. The research team is working to get the required paperwork and website set up to support the start of the 4th round of research. The official announcement will occur shortly.

All GF members could support this effort by encouraging the professionals from their metabolic clinic to consider become peer reviewers for the research team. Each proposal is assigned two peer reviewers to evaluate the proposal to ensure it utilizes the best science. Please contact Dan Lambert,

research@galactosemia.org if you know any professionals (physicians, researchers, dieticians, speech therapists, etc) that might be interested.

The research team welcomes three new members Christine Winey, Kayla Costner, and Beatrice Ortego. They join Brian Mannix, Christy Johnson, Denise Wilburn, Jo Beth Southard and Kristine Saylor. Dr. Mark Winey, a Genetics Professor at the University of Colorado, Boulder is also helping the research team in identifying peer reviewers for supporting the 4th round of research.

Meet Miracle Child and Enid Native, JASE!



Jase stunned the world with a beautiful head of dark hair upon his birth on August 18, 2010. This healthy baby was able to return home with his parents despite a touch of jaundice. Over the course of several days, his jaundice worsened, he began losing more weight than normal, and he turned an odd shade of grey. At six days old, Jase's newborn screening revealed that he had galactosemia, a metabolic disorder characterized with an enzyme deficiency that makes it difficult to metabolize galactose, the main sugar in milk. If the body is unable to metabolize galactose, it can become toxic to the liver. Due to his mother's breast milk, Jase became severely dehydrated and lost 13% of his birth weight. After switching to strictly soy milk, the following weeks were a rollercoaster ride as Jase would improve one day and decline the next. Today, he is unable to eat anything containing lactose or galactose and has to endure blood tests every six weeks. With the help of Dr. Klaas J. Wierenga and a metabolic dietician, Jase maintains his levels with a restrictive diet. He has begun speech therapy to help with his speech delay, caused by galactosemia. He loves to wrestle, admire the monkeys at the zoo and has big dreams of eating dairy-free mashed potatoes at every meal.

"Jase came to my attention first as a result of an abnormal newborn screening. The genetics department takes part in the majority of the kids that have abnormal results on the metabolic screening. If galactosemia is not detected at birth or shortly thereafter, it can actually become fatal to the child. Thanks to the newborn screening and a strict diet, Jase is doing extremely well!" – Dr. Klaas J. Wierenga

RACE 4 JASE
5K Run & 1 Mile Walk
All proceeds will benefit the
Galactosemia Foundation
Saturday, April 20, 2013, 8am
Crosslin Park, Enid, OK

FUNDRAISING

After our 2012 Conference the fundraising team has heard from many of you interested in fundraising for the first time! We are so excited to have so many new families interested in helping the Galactosemia Foundation do good work! Thank you! We can't wait to hear about your fundraising adventures.

The aunt of Sydney Miller has a talent for writing grants. She put her talent to use for the Galactosemia Foundation by writing a grant she found from the Build-a-Bear Corporation. We are thankful to Cari Miller for using her talents to afford us the possibility of grant money! Thank you Cari!

Also, thanks to Scott Shepard, our Good Search account now is accurate with our new name! You can use goodsearch.com for all your internet searching needs and GF will reap the benefits. We have raised almost \$1200 so far just using this search engine! If you are not already taking advantage of this fundraiser, please go to goodsearch.com and type in "Galactosemia Foundation" as your cause and watch those funds rack up!

If you are planning a fundraiser PLEASE keep us informed. We would like to know what you are doing and where so we can support you and thank you properly! E-mail any fundraising information to fundraising@galactosemia.org.

The fundraising team is looking to expand and organize! If you are interested in working on this team, please e-mail us at fundraising@galactosemia.org. We need people to keep track of fundraisers, write thank you cards, etc.

The fundraising team would like to encourage all families to think about how you can make a difference. If you need any help or encouragement, please contact Denise Wilburn at fundraising@galactosemia.org. If you would like to view fundraising ideas or how-to's visit the Galactosemia Association of Midwest America's website at galactosemiamidwest.com and select "fundraising."

No Milk For Joseph Fundraiser

A spaghetti dinner will be held on Friday, March 8, 2012 in Troy, New York to help raise funds for Galactosemia Foundation. "No Milk for Joseph" Tickets are \$10. It's being held at the Italian community center. 1450 5th Ave. Troy NY 12180. 11am-8pm.

The website for this event is NoMilkForJoseph.com where people can pre-order food, shirts, etc.

The Galactosemia Foundation invites you to support fundraising for the 2014 Orlando Conference by shopping at any Kroger store in Louisiana and Texas and participating in the Kroger Neighbor to Neighbor Donation Program. All you need to do is go the customer service booth and ask the representative to do a one-time scan of the attached customer letter to your KrogerPlus Card which allows you to participate, at no cost to you, in raising funds for Galactosemia Foundation through grocery shopping or the purchase of gift cards.

The Galactosemia Foundation is also participating in the Randalls/Tom Thumb/Simon David Good Neighbor Program and our organization number is 11975. You only need to request that the Courtesy Booth link your Remarkable/Reward Card to our fundraising number. This program is available on a state-by-state basis.

We request that you please share this information with all your friends, family, employees or co-workers, research groups, email lists, blog sites or organizations you can share this information with! Also, if any of the membership knows of any additional promotional gift card programs or charity fundraising programs which raises funds for charity organizations, please contact Sue Smith at 281.381.5091 and she will be happy to take your information and contact them.

We are already hard at work on the 2014 Orlando Conference and your participation in fundraising will assist us in achieving our goal of providing a low-cost, high-quality 2014 conference!



Customer Letter

Step One:

Read through this letter to understand the advantage of being a part of the Kroger Neighbor to Neighbor Donation Program.

Step Two:

Take the letter with you to your neighborhood Kroger store the next time you go shopping.

Step Three:

Present the letter with your organization's barcode to your cashier upon checkout. After they have scanned your KrogerPlus Card you will be enrolled for the current year of the Kroger Neighbor to Neighbor Donation Program. Every time you shop at Kroger and use your enrolled KrogerPlus Card, Kroger will contribute a percentage of your eligible purchases to the Kroger Neighbor to Neighbor Donation fund. Once a card is scanned with the barcode, it will be active for the remainder of the program year. After 7 days, from initial scan, you will start to see your contributions to your organization on your receipt.

Q & A

How many Kroger Neighbor to Neighbor accounts can an organization have?

One per organization (Example: ABC HIGH SCHOOL account could represent: PTA, cheerleaders, football, basketball, band, etc.) Unless each entity has their own 501(c) (3).

Can a household "link" their KrogerPlus Card to more than one organization at a time?

No, the system allows for one organization per household. To change organizations, you must present your KrogerPlus Card and the new organization's barcode to the cashier.

Do I have to enroll each program year?

Yes, all organization's and participants must re-enroll for each new enrollment period. The current Neighbor to Neighbor Program is accumulating period is effective **July 1, 2012** to **April 30, 2013**. Organization Enrollment period is **June 1, 2012** to **Dec. 31, 2012**.

How much can my organization earn?

The Kroger Neighbor to Neighbor Donation Program will donate \$1 million annually. Each organization will earn a percentage of the \$1 million equal to the percentage of total earned contributions attributable to that organization.

How are the funds distributed?

Contributions will be paid via check and mailed to the organization's address and primary contact identified on the organization application form. For schools, it will be mailed to the principal and the funds distributed at their discretion.

GALACTOSEMIC FOUNDATION



Kroger cashier: Please scan customer's KrogerPlus Card at the beginning of the order, and then scan the above barcode. The customer's KrogerPlus Card is now enrolled in the Kroger Neighbor to Neighbor Donation Program.

Please feel free to contact a Kroger Neighbor to Neighbor Donation Program Representative with any additional questions at 866-995-7643 or email to neighbortoneighbor@kroger.com. Additional information is also available at www.krogerneighbortoneighbor.com

Note on Golf Outing:

The Saylor and Rodger's family will be hosting the 5th annual "Fore the Cause" golf outing to benefit the Galactosemia Foundation. This year's event will be held Friday May 17th, 2013 in Chesterfield, Va. Last year's event raised over \$40,000. If you live in the Richmond, Va area and would like to attend or would like more information please email ssaylor97@verizon.net. We will send out more information in the next newsletter.

Raise money for Galactosemia Foundation

by searching the internet, shopping, and dining

How many times do you and your family members search the internet each day? How much money do you spend buying stuff over the internet or going out to eat during a year? Just imagine if the Galactosemia Foundation could receive \$0.01 for every search, up to \$0.30 for every dollar you spend shopping online, and up to \$0.06 for every dollar you spend eating out? Well, stop imagining and go to www.goodsearch.com! You are already searching, shopping, and eating so why wouldn't you use GoodSearch, GoodShop, and GoodDining to benefit Galactosemia Foundation?

Funding is vital for the Galactosemia Foundation to continue our mission to educate, support and provide advocacy for those affected by Galactosemia (and to pay for things like printing and mailing this newsletter). Galactosemia Foundation has received about \$200 per year in donations from GoodSearch (\$1150 total since April 2006) with just a few active users. With your help, we could easily increase this amount by 10x to \$2000 each year!

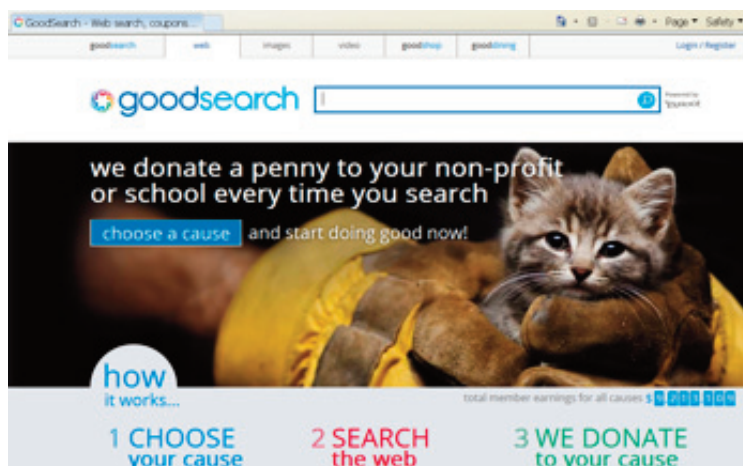
What are GoodSearch, GoodShop, and GoodDining?

GoodSearch is a search engine (powered by Yahoo) that donates 50% of the revenue generated from sponsored search advertisers to charities. This works out to about \$0.01 for every search. GoodShop is a portal to well known online retailers (2,500+ retailers, including Amazon, iTunes, eBay, Groupon, Overstock.com, travel sites like Priceline, Orbitz, etc, retail stores like Target, Bed Bath & Beyond, Walmart, etc.). Plus, give back through GoodShop and you can save with over 100,000 coupons. You simply click the link from the GoodShop site to the retailer and then shop like you normally would except Galactosemia Foundation gets a donation. GoodDining is a program that includes over 10,000 restaurants throughout the United States and Canada that donate a percentage of your purchases.

How do you GoodSearch, GoodShop, and GoodDine?

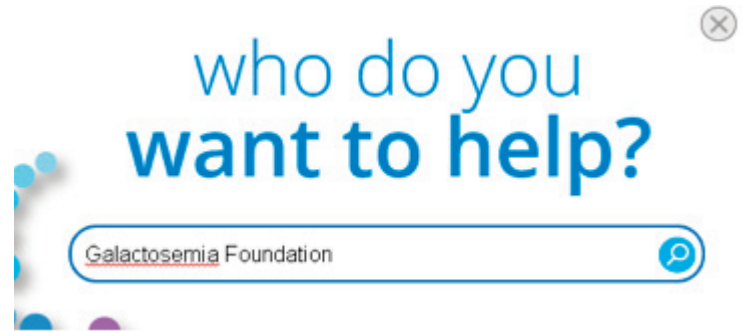
Galactosemia Foundation is already set up as a charity, so it is easy.

1. Go to www.goodsearch.com

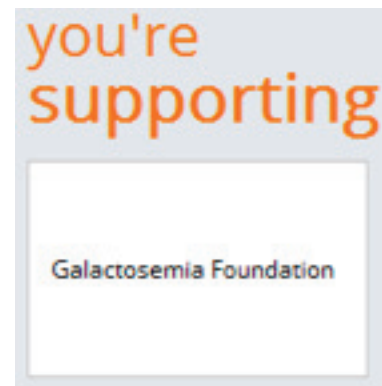


2. Optional: click 'Register' at the top right to create an account. With an account, you can keep track of how much you have raised.

3. Choose your cause (search for Galactosemia Foundation).



4. Please make sure you are supporting Galactosemia Foundation each time you search



- a. If you have registered and selected Galactosemia Foundation as your charity, then just make sure that you are logged in.
- b. If you are not registered and logged in, then you have two options:
 - i. If you allow "cookies", the last nonprofit you designated will remain chosen. If you or your anti-virus software deletes your cookies your searches may not benefit Galactosemia Foundation!
 - ii. Another option is to link to GoodSearch with the following direct link, which includes Galactosemia Foundation's Charity ID and bypasses the "cookie": <http://www.goodsearch.com/?charityid=806926>

Now you are ready to raise money for Galactosemia Foundation by searching the internet and making online purchases. You have to enroll separately for GoodDining (and register a credit or debit card) – see <http://www.goodsearch.com/howgooddiningworks> for more details.

Here are several suggestions to help us raise the most money:

- Set <http://www.goodsearch.com/?charityid=806926> as your home page on all of your family's computers and smart phones at home and at work.
- Click <http://www.goodsearch.com/getinvolved.aspx> for other suggestions about how to spread the word about supporting Galactosemia Foundation through GoodSearch (such as by adding a search box to your websites, adding a footer to your emails, etcetera).
- Please do not abuse GoodSearch (repeated manual clicks, the use of robots or other automated query tools, etc) or else you could be banned and/or Galactosemia Foundation could be delisted.

Holiday Shopping



The Holiday Season is fast approaching. Help support the Galactosemia Foundation while doing your Christmas shopping this year. You can support in many ways... Goodshop, purchase our pearl Swarovski crystal necklace, purchase our Swarovski crystal bracelet, order a customized, bling, electronic case or simply donate funds in someone's honor.

Bling for a Cause!

Turn you cell phone, ipad, kindle, or any electronic case into a custom, embellished work of art and support a great cause at the same time. Tori Fowler, sister of galactosemic Landon Fowler, is designing cases upon special request to fit your style and budget with 100% of the proceeds going towards The Galactosemia Foundation. Please contact Tori today via email at blingforacause@galactosemia.org to get started with details of your order. Thank you for helping to support galactosemia!



Swarovski crystal necklace and/or bracelet

These very unique pieces are loved by all and also make wonderful presents. These pieces tell a heartwarming story of galactosemia and serve as a reminder to the person whom is wearing the piece.

GoodSearch Contest Announced

GoodSearch Contest Dates

December 1, 2012 – February 28, 2013



Concept

The three people that earn the most money between December 1 and February 28 using GoodSearch, GoodShop and/or GoodDining on behalf of "Galactosemia Foundation" will win some Galactosemia Foundation merchandise. Winners will be announced Mar. 3, 2013.

Prizes

Prizes will include Galactosemia Foundation merchandise, such as shirts, bracelets, magnets, and other such items.

Instructions

To enter the contest, visit www.GoodSearch.com, register with a valid email address and list "Galactosemia Foundation" as your cause. You will be able to access GoodSearch, GoodShop and GoodDining through this same website for the duration of the contest. Make sure you are logged into GoodSearch with your username each time you search and make purchases in order for them to be credited to your account.

To earn donations through GoodShop, search for a store at www.GoodShop.com and click "Shop this store now." You will be directed to that store's website and a specified percentage of your purchase will go to Snowbird. You must access the online vendor through GoodShop in order for your purchase to benefit Snowbird. GoodDining requires additional registering at www.GoodDining.com. Please review complete instructions on the website.

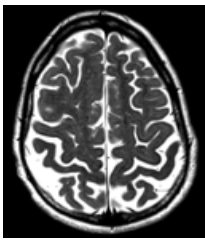
Disclaimers

** All participants, whether newly registered or veteran GoodSearch users, will be assumed to start the contest at \$0.00 on December 1 at 12:01 a.m.

** GoodShop donations from purchases may take up to eight weeks to clear and be credited to your GoodSearch account.

** Amazon.com purchases via GoodShop benefit Galactosemia Foundation but are not reflected on individual accounts. Money earned through Amazon.com will go into Galactosemia Foundation's general fund raising account.

For questions or to request additional information, please contact Scott Shepard, Galactosemia Foundation Vice President, by email at vicepresident@galactosemia.com.



The Brain and Galactosemia: A pilot study

The Galactosemia Foundation provided much appreciated support to Boston Children's Hospital and the Beth Israel Deaconess Medical Center for a study on the Brain and Neuro-functioning in Adults with Galactosemia. One of the most vexing questions in galactosemia is why some individuals experience significant problems while others evidence difficulties to only a mild degree or not at all. Advanced neuroimaging (magnetic resonance imaging or MRI) and EEG studies in combination with neurological and neuropsychological assessments have the potential to reveal basic information about the origin of developmental, cognitive, and neurological issues in galactosemia.

In a previous study, also funded by the Galactosemia Foundation, 33 adults came to Boston for an amazing weekend of conversation, group activities and 12 separate research evaluations, including medical, psychological, speech, bone density, endocrine, neurological and nutrition assessments. Three of the main conclusions were:

- There is a clearly defined neurological phenotype in Galactosemia. This means that we found a typical pattern of cognitive strengths and weaknesses and neurological signs among the adults we studied.
- From a genetics point of view, there is variable expressivity in the brain. This means that some people had severe symptoms while others were unaffected, even when the gene mutations for galactosemia were the same.
- The right hemisphere of the brain is selectively vulnerable. This conclusion was based on the types of problems noted among the adults with galactosemia. Motor coordination problems, tremor, depression, anxiety, and difficulties with map reading and finding one's way are considered signs of right hemisphere abnormalities.

Based on these conclusions, we wanted to find out more about the brain in galactosemia. The current study had 4 aims:

1. Conduct a comprehensive neurological examination focused on neurological abnormalities related to congenital developmental asymmetry in the brain
2. Conduct a comprehensive neuropsychological examination focused on a comparison between right and left hemispheric functions including visual-spatial problems, reaction time, language processing issues and neuropsychiatric manifestation
3. Conduct an EEG to examine regional differences and asymmetries in frequency analysis

4. Conduct detailed MRI examinations to obtain volumetric measurements of the brain and diffusion tensor imaging (DTI) to examine white matter neuropathways

Five men and five women participated (average age 27.5 years). Each had a thorough neuropsychological and neurological evaluation plus an EEG and 9 subjects had an MRI involving advanced methods for calculating not only the size of different parts of the brain, but the density of the neuropathway fibers (that control how efficiently or quickly messages can go from one part of the brain to another). Analyses of all the data are not quite finished, but preliminary results are described below:

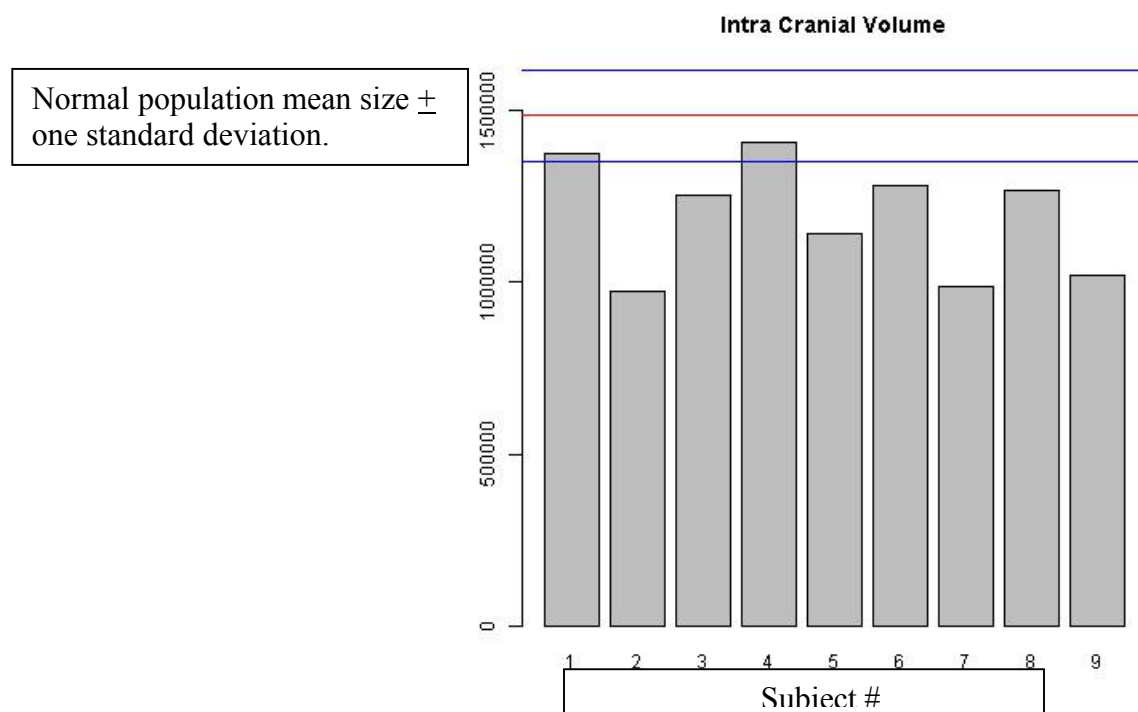
IQ ranged from 51 to 108 with an average score of 80. (The average range in the general population is 85-115). The neuropsychological evaluations indicated a similar pattern of strengths and weaknesses among the entire group. Almost all the adults did better on the verbal subtests (vocabulary, verbal reasoning, knowledge of the world) than on the nonverbal subtests. Weaknesses were noted in visual motor coordination, visual perception, auditory processing, and visual-motor speed. Five subjects rated themselves as anxious and 2 rated themselves as depressed.

The neurological evaluations also revealed a range of presentations, varying from normal to subtle right hemisphere abnormalities. Very few subjects had bihemispheric dysfunction (right and left brain difficulties). Other common findings included mild scoliosis (crooked backbone) and loose joints. The muscles in the mouth were normal. Very few of the adults had dysarthria (slurred speech) and none exhibited apraxia of speech (disjointed speech). However, most showed reduced prosody (which means they tended to speak in a monotone). Six of the 10 subjects had some form of tremor, 4 had trouble with balance, and 2 had dystonia (abnormal posturing).

Consistent with the neuropsychological and neurological findings, 6 of the 10 adults showed a mildly abnormal EEG, with 5 of those having right hemisphere abnormalities and 1 subject having abnormalities in both the right and left hemisphere.

Nine subjects received the MRI. (One subject could not tolerate being in the closed machine and had to stop.) Results showed that 7 adults had what are called T2 White matter signal abnormalities. This means that the white matter (myelin) is abnormal, usually signifying slower and less efficient cognitive functioning (and some motor difficulties). The MRI also showed that specific areas of the brain, usually associated with motor functioning, were smaller or had abnormal white matter. The brain ventricles (open spaces) were larger in the front of the brain, suggesting more frontal atrophy (loss of function). The ventricles enlarge when there is cortical or white matter atrophy. There were right-left brain differences, but these need to be analyzed further to understand their meaning. As noted in the figure below, 7 of 9 subjects had a brain volume below the normal population mean. Brain volume correlated with IQ ($p = .0003$). Subjects 1 and 4 had the highest IQ scores, normal EEG and normal MRI findings. Subjects 3 and 7 were having the most difficulties in all respects. Two subjects had previous MRI results from 10 years ago that could be compared to current findings. In one subject, the MRI

findings were the same. In the subject with the most serious clinical presentation, there was evidence of progression.



So, what does all this mean for adults with galactosemia and parents?

Adults, ask your clinicians to provide written instructions since auditory processing and memory tasks are difficult. Ask your clinician to give you plenty of time to respond to questions. Notice if you are having signs of depression and anxiety. Medications for anxiety and depression usually help adults with galactosemia. Know that finding your way may be challenging, so use a GPS and be patient with yourself if you get lost on the way.

Parents should be sure to obtain a developmental evaluation for the child and start early intervention services, if indicated, since the MRI results suggest that changes to the brain occur early. Don't be surprised if tremor develops or if the child experiences difficulties with motor coordination and motor planning, especially if an MRI shows basal ganglia abnormalities. Moreover, if MRI shows T2 white matter signal abnormalities, address potential learning disabilities through special education services in the schools.

We concluded that neuropsychological evaluations, neurological evaluations and MRI studies are useful for baseline as well as clinical purposes. If an MRI cannot be obtained, an EEG may serve the purpose of identifying individuals with right brain vulnerabilities associated with the common difficulties noted in galactosemia.

Our next step will be to obtain funds to study other genes in individuals with galactosemia that might predict who will have difficulties in what areas. We also hope to conduct a larger study using advanced MRI methods to replicate our findings from this small study and associate results with other genetic and biochemical findings. So, stay tuned and thank you to the subjects, their families and the Galactosemia Foundation!

Susan Waisbren, PhD
Penny Greenstein, MD
Gerry Berry, MD

(With additional thanks to a wonderful team: Donald Schomer, MD, Ellen Grant, MD, Sanjay Prabhu, MD, Katyucia Rodrigues, MD, Vera Anastasoae, Stephanie Petrides, Matthew Brown)

Dear Galactosemia Foundation Families:

First, a huge **THANK YOU THANK YOU THANK YOU** to the conference organizers and to all the wonderful volunteers and families of the Galactosemia Foundation for once again welcoming us into your "extended family" in Dallas last July. You all are the BEST!

Our research is moving along nicely and as we finalize and publish further results I will be delighted to share these findings with you.

We are also gearing up to initiate a new arm of our study – looking at **gut microbiota** (from stool samples... yes, you read that correctly) in children with classic galactosemia. This is no joke, but an important new area of research and intervention in many health fields, and it is about time we applied this new approach to see if abnormal gut microbiota or other gut issues might be causing some of the problems experienced by people with galactosemia. So... please watch for future emails from me as we will soon be recruiting volunteers for this new arm of our study. Interested? Please let me know (jfridov@emory.edu or 404-727-3924), and thanks!

Finally, Dr. Nancy Potter and I have been discussing ideas for possible new joint projects so please stay tuned for more information!

Thank you!



Judith L. Fridovich-Keil, PhD
Professor, Department of Human Genetics
Emory University School of Medicine

Thank you
Todah Xie Xie
Mahalo Spacibo
Salamat Merci
Arigato
Khawp khun
Obrigado
Danke Gracias

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 | |
|---------|-------------------------|----------|----------------------------------|-----------|------------------------------|
| | | | | 8 | No Milk For Joseph, Troy, NY |
| APRIL | | MAY | | JUNE | |
| 20 | Race For Jace, Enid, OK | 17 | Fore The Cause, Chesterfield, VA | | |
| JULY | | AUGUST | | SEPTEMBER | |
| | | | | | |
| OCTOBER | | NOVEMBER | | DECEMBER | |
| | | | | | |



**Galactosemia
Foundation**
Linked for Life.

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

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

