

# The Galactosemia Gazette

Volume 2, Issue 1

Fall 2006

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## **PGC Conference 2006**

*bigger than ever and something for everyone*

PGC held its 2006 Conference in Philadelphia, PA on July 28th and 29th. The conference included an adult program with nearly 300 attendees in the main ballroom, a youth program with nearly 100 kids aged 5 to 17, a childcare program for children under 5, and a *Generation G* program for young adults with galactosemia (see article inside).

Presentations in the main ballroom included talks on diet, research, speech and language, neuroimaging, lactose-free medications, learning disorders, regional group updates, and an 'ask-the-experts' panel discussion.

Breakout topics included diet from birth to 5 years, diet in the school age years, diet in the adolescent years, topics in special education, galactosemia-101, special needs financial planning, premature ovarian failure, occupational therapy, speech—beginning talkers, speech—school age, and 'ask-the-parents' panels.



In addition to the daytime presentations, there was a dessert mixer on the first evening and an outing to a minor league baseball game on the second evening. Conference attendees received a T-shirt, tote bag, recipe book, roster of attendees, Doctors list, and 'Our Stories' booklet.

The PGC community owes a great debt of gratitude to Angela Forsythe and Michelle Fowler who worked so tirelessly to put on this conference. Folks are already looking forward to 2008—stay tuned!

## **Help PGC While You Surf the Web**

Helping PGC raise funds has just gotten easier. The website [www.goodsearch.com](http://www.goodsearch.com) is a search engine (like yahoo or google) that donates 1 cent to the charity of your choice for each search done on their site. If you go to the [www.goodsearch.com](http://www.goodsearch.com) website, you will see a box in the center of the screen near the words "I'm Supporting", just type

### **Parents of Galactosemic Children**

in this box, then click on the 'verify' button. You will see

Parents of Galactosemic Children  
(Gautier, MS)

appear in the box.

Your web browser should remember PGC as your favorite charity each time you come to the goodsearch website. Then you can type your search topic in the regular box at the top of the page. Each time you do, PGC gets another penny!

This simple fundraising approach can be very lucrative. The autism charity sponsored by American Idol star Clay Aiken is raising over \$300 per month! So please spread the word about this site to your family and friends.

## Wrap-up Report From PGC's 2006 Conference Committee

The theme of PGC's 2006 conference was, "Promising, Gratifying, Collaboration," and the conference was a wonderful experience for many families. The Philadelphia location allowed many families to combine a summer vacation with the meeting and enjoy a bit of history during their stay. We are pleased to say that PGC offered something for everyone this year. Adults had their own session while children played and napped, youth were busy learning and doing different projects and crafts while our "young adult" galactosemics had their own program enjoying things from lunch with doctors to parent-sponsored offsite activities.

The main adult program was a combination of general sessions and breakouts during the two full days. There were a total of 12 general sessions and 14 breakout topics. Of course, the success of the educational sessions can only be achieved through the generosity of the professionals and PGC "family members" who donated their expertise. The collective experience of the presenters was quite impressive, with five physicians, three Ph.D.'s, a pharmacist, special education teacher, attorney, dietician, occupational therapist, and financial planner. There were also over ten speakers who shared their personal experiences with galactosemia that certainly added to the richness of the program.

Friday started off with Dr. Judy Fridovich-Keil explaining her current research project. Families also had the opportunity to participate in this research onsite at the conference. Next was Laurie Bernstein, a PGC family favorite. Laurie once again delivered much useful information on the Galactosemic diet.

Dr. Berry gave us a very scientific, but informative overview of galactosemia and expressed to us the need for further research. The highlight of Dr. Berry's talk was to follow when Michelle Fowler, PGC President, and Sarah Roth from Children's Hospital in Boston, announced Dr. Berry's relocation to Boston and the opening of the first ever, Galactosemia Center of Excellence. As a result of the unprecedented fundraising success of Kristine and Sean Lydon's "Kyleigh's Cure" organization (see story page 4), PGC was able to donate \$70,000.00 to establish this center and we are committed to fundraising in order to make future contributions to the center in order to support its growth and success.

Next up was Dr. Andrew Newberg, an expert in functional MRI and PET scanning from the University of Pennsylvania. After a delicious galactosemia-safe lunch, PGC's "personal pharmacist" (we can say this, because he calls himself that !!), William Bell, updated us on pharmacy issues and medication.

We then heard from a panel of 'galactosemic adults' sharing their stories about growing up and their personal stories of living with galactosemia. Ray Mansfield, Sharon Howell, and Maureen Bell all had touching stories and we appreciate that they shared them with all of us.

Our afternoon finished out with a number of breakout sessions covering topics for new parents, special education, diet, research, financial planning and networking.

Wrapping the evening up was a 'dessert social' complete with a wonderful cake donated by the GANES group that was enjoyed by everyone. The cake was made by Angela Forsyth's mom especially for our group and was, of course, 'safe' for the Galactosemic diet.

Saturday started bright and early with Dr. Elsas's discussion of various topics in galactosemia. Dr. Elsas is also doing galactosemia research, and families had the opportunity to participate in his study at the conference.

Dr. Selznick, a learning specialist from the area, was up next. He spoke to the general session regarding learning disabilities and strategies for helping struggling learners.

We were pleased to have Dr. Nancy Potter in attendance. A handful of families got to meet Dr. Potter this past year as she did a research project on the speech of those with galactosemia. It was fascinating to hear Dr. Potter describe her research findings.

The morning session ended with regional updates from around the country. There are currently three local nonprofits in existence and more on the way! Michelle Fowler, filling in for Beate Krull whom was unable to attend, announced a new group that has been formed in the Northwest; Galactosemia Support Northwest, and gave some details of the

group. Barb Bense represented the GFMN group; Galactosemic Families of Minnesota, and gave a progress report on their activities. Last, but surely not least, Angie Forsyth gave an update from the GANES group; (Galactosemia Association of the North Eastern States).

The afternoon started off with breakout sessions. Many topics were offered, from diet to speech to POF to occupational therapy. Attendees were able to choose two topics. The last main session of the day was the popular "Ask the Expert Panel." This was a great forum to challenge those experts and ask those questions that we all have.

Overall, this conference was rated 'very good' by the attendees on the conference surveys that were filled out. Families were able to share stories and experiences with others. It was nice to be in a place where everyone you were around actually understood what you meant when you said 'galactosemia'.

For both of us, it was such a wonderful experience seeing the children's faces light up in knowing there were others in the world with galactosemia just like them. Also, galactosemic children and adults being able to safely eat from the 'safe' buffet lunch and snacks brought a smile to our faces!

Thanks to everyone who attended and to all of our wonderful speakers and volunteers. This would not have been possible without you! We were also fortunate to have corporate sponsors and donations to help with the meeting. Thanks again to our generous sponsors:

Amanda's Own Confections  
Chocolate Emporium  
GANES – Galactosemia Assn. of the North Eastern States  
Illinois Nut Company  
Pin Pro Promotions  
Turtle Mountain

Lastly, conferences such as this rely entirely on volunteers from our PGC family. The conference has grown by leaps and bounds in terms of what is being offered and the amount of people attending. Conferences will not be possible in the future without the commitment of volunteers to make it happen! There are many ways to get involved and many areas to choose from. We encourage you to contact PGC and volunteer to help with the next conference – you can donate anywhere from an hour or two to ... well ... as much as you like!! If we all pull together, just imagine what we can accomplish!

Sincerely,  
Angie Forsyth – 2006 Conference Coordinator  
Michelle Fowler – PGC President

## ***PGC Logo Items available***

Did you miss your chance to pick up some fun PGC logo items at the conference? Fear not, many items are still available for purchase. We have:

Aqua Tee's (Adult: S,M,L,XL,XXL) - \$12.00  
Conference Tee's ( Youth S, Adult S,M,XL,XXL) - \$10.00  
Ornaments — \$10.00  
Back Packs — \$6.00  
Tote Bags — \$8.00  
Huggers — \$2.00  
Lunch Bags — \$10.00

See the PGC website in coming days for pictures of these items.

Contact Michelle Fowler by email at [president@galactosemia.org](mailto:president@galactosemia.org) or by phone a 1-866-900-7421 to place an order.

# Amazing Fundraising Success !!

*Editors note - The Lydons told the story of their daughter Kyleigh's birth and diagnosis and some of their initial fundraising activities in the last PGC newsletter. In that article, they concluded by describing how they would be meeting with some close friends in order to come up with more fundraising ideas. This article tells of the astounding success that came from their hard work and dedication since then. The entire PGC community is blessed to have the talents of this wonderful family to benefit us all.*

One night in August of 2005 we had gone out to dinner with six of our closest friends. We shared our feelings with them about having some sort of fund-raiser to raise money for galactosemia research. For the remainder of the evening everyone came up with different ideas for a fund-raiser and we all decided that a live/silent auction, if planned correctly, would raise the most money. Our main goal was to raise as much money as we could. From here we divided our tasks and set up many meetings. At first, everyone was so enthusiastic to help. Along with planning the live/silent auction, we also did other things to raise money such as selling candy, and galactosemia awareness bracelets. Also, our friend raised money by getting people to sponsor her for running the Chicago Marathon.

Some of our first tasks were to name our organization (which we decided would be "Kyleigh's Cure"), to open a benefit fund bank account, to obtain a tax I.D. number, and to have informational flyers made up to be mailed out and delivered to businesses. We also eventually created a website which is [www.kyleighscure.com](http://www.kyleighscure.com). The original group of friends who began planning this event with us was named the "Kyleigh's Cure Committee". The next steps were picking a date, location, and hitting the pavement to look for donations. We found that people who we expected to help didn't while others who didn't even know us donated generously once they heard Kyleigh's story.

In the middle of all this planning we were also expecting another child and were both working full time jobs on different shifts with different days off. As the days went by, we started to feel so overwhelmed and felt as if we had bitten off more than we could chew. It always seemed that at that moment Kyleigh would smile or do something funny. Just looking at her and knowing that what we were doing might help her kept us going.

As the date came closer, this event began to snowball. Our house was taken over by all the items donated. More and more people wanted to help. We originally had two hundred tickets made up and distributed them to all of our friends and family to sell. The only problem was that the tickets were not selling. There were so many different components involved with organizing this event; it just seemed as if the pieces were not coming together and we started to get very nervous. We have a friend who is a professional auctioneer and is also very active in fund-raising events. We called him up one night and he came to the very next meeting. All of our friends loved him. He gave us many helpful ideas, and pointed us in the right direction to make this all come together. He also agreed to be the auctioneer for our event.

Another of our friends contacted a journalist from a local newspaper who contacted us and wanted to meet with us. We were interviewed at our house and she wrote a beautiful article about Kyleigh's story and the event we were planning. She also told us that it was highly unusual for a family to raise money for medical research and not for themselves. Immediately after the article was published we began receiving donations and beautiful letters wishing us luck from people who read the article. People were also requesting tickets to the event. After the first article was published, two other newspapers contacted us, one of which was the Boston Globe, (a paper sold nationally). Articles were written in both. The second two articles generated even more donations and requests for tickets.

Oh, we certainly can't leave this part of our story out. On New Year's Day, 2006, I was rushed to the hospital and had an emergency c-section one month early. We were blessed with a healthy beautiful baby girl who, after looking through the baby name book in the recovery room, we named Samantha Rose. We were working so hard on the fund-raiser, and thought we had another month to come up with a name, pack a suitcase, buy a crib, and find someone to watch Kyleigh and our two dogs, Zack and Lou. Once we got home from the hospital, we knew we had no time for recovery or adjusting, we had to jump right back into the planning of the fund-raiser as the date was fast approaching. After convincing the doctor to let me leave the hospital a day early, we had to agree to have a visiting nurse come to the house the next day. When the nurse came to our house, we could sense her tension with the chaotic atmosphere of our house, one new baby, another angry baby because of the new baby, two barking dogs, and the house filled with items for the fund-raiser piled to the ceilings. She suggested that we call a local church and ask for people to volunteer to help with the "out of control" situation in our house. Needless to say, after this suggestion, she was shuffled right out the door because we had work to do.

The week of the event was absolutely crazy. Our entire house was filled with donated items, and boxes of gift certificates, all of which had to be separated and put into baskets for the silent auction. This was such an unbelievable amount of work. It took 15 people two nights to make these auction baskets; all the while more donations and items were still coming in. We ended up having over 70 beautiful baskets. For the live auction, family and friends donated trips, time share vacations, catered dinners, sports memorabilia, tickets to events, sports games, and autographed items from musicians including a guitar signed by the country singers Brooks & Dunn.

The ticket sales started to pick up a couple weeks before the event and we were shocked to realize that we had to order a couple hundred more tickets. All along we were trying to figure out how many people would come to our event. We would have been happy with two hundred people. When we learned on the day before the benefit that our final ticket sales number was 425, we were in shock. People continued to call looking for tickets the day of the event so we had a sign-in table and had to have someone sit there to sell tickets (even though we had been told that we could not sell them at the door). We still don't know the exact number of attendees; but it was well over 500.

In addition to the auctions on the night of the event, we held two 50/50 cash raffles, one of which was won by our boss. He won \$1250, and gave it back to us to put towards the research. We also had a game called 'heads or tails'. We purchased several hundred blinking pins for the heads or tails game, and sold each pin for 20 dollars. The way this game worked was everyone with a pin was called out on to the dance floor, at which point they were told to put their hands on their head or on their tails, at which point a coin was flipped. Those who correctly guessed which side the coin landed on could continue to play while those who guessed wrong were asked to take their seats. The game continued until one person was left standing. The prize for this game was a night at the Four Season's Hotel in Boston with deluxe accommodations, dinner at Maggiano's restaurant, and dessert at Finale's in Boston. This game raised over \$3000.

The night went off without a hitch. It was such a wonderful night and we got so many compliments. All of the hard work paid off. At the end of the night, after cleaning up, we were sitting around a table with a group of close friends, and we were all trying to guess what the final amount raised was. The highest guess was \$50,000. We were floored that this might have been possible. The next day, it took us all day to count the money, and figure out the credit card receipts, and count donations that were made that night at the check-in table. We came up with a total of over \$83,000, and we immediately began counting this again as we knew this couldn't be possible. We were wrong, and the second time around we came up with over \$85,000 as we had originally added the credit card receipts incorrectly. We were all crying and couldn't believe it. Donations continued to come in and our total fund-raising amount (2005 and 2006) for PGC was approximately \$93,000.00.

Although this was extremely stressful and time consuming (and we still have over 2000 people and businesses to send thank-you notes to), it was all worth every bit of effort. The day of the event we promised that we would never do this again. But the day after, we were so happy that we were already talking about fund raising ideas for the future.

Our point of explaining every detail of the hard work that went into raising this amount of money is to show that we are just two average people who are no different from anyone that was at the conference in Philadelphia. We don't come from money, and had many odds against us. If we could accomplish this, anyone could. We are just two people that are very driven and want a better quality of life for our daughter Kyleigh and all the other children with this disorder.

Are you tired of medical professionals not having answers to your questions? Are you tired of the inconsistencies in the diet recommendations and the variations in different clinics due to the lack of research? If we want things to change we need to raise money for research. If we don't raise money the research will stop.

We know that our contribution was just enough to get the center of excellence off the ground and the PGC needs to raise more money in the future to continue the progress. We realize that not everyone would be willing to sacrifice as much as we did to raise this money, however, we have many wonderful and simple ideas to raise money that we are eager to share. We can all work together and make this happen.

Sincerely,  
Sean & Kristine Lydon

## Center of Excellence for Galactosemia Research Announced

In August 2006, PGC announced its gift of \$70,000 to support Children's Hospital Boston's plan to create a model Galactosemia Center which will be led by Gerard T. Berry, MD, a renowned galactosemia specialist. As a primary teaching hospital of Harvard Medical School, the center at Children's will feature state-of-the-art diagnostic and treatment facilities, an intensive basic and clinical research agenda and varied educational programs and materials for doctors-in-training, practicing physicians, patients and families. Once established, the center will be a distinct entity in the landscape of galactosemia care, research and training.

The Center will build on Children's long tradition of innovation in galactosemia care and study that began in the 1970s, when Harvey Levy, MD, and his colleagues conducted a number of seminal galactosemia studies. For example:

- They were the first to identify speech and language deficits among children with galactosemia.
- They identified overwhelming infection as a major cause of death in babies with galactosemia, leading to early recognition and lifesaving treatment.
- They identified the buildup of toxic metabolites in the fetus with galactosemia despite milk avoidance during pregnancy, leading to the discontinuation of this dangerous practice.

Much more can be done to build on this groundbreaking work. Children's Hospital's expansive pediatric research enterprise – the largest of its kind in the U.S. – offers an ideal setting to advance galactosemia research. The center will extend its capabilities well beyond Children's Hospital, spurring local and national outreach initiatives that engage teachers, guidance counselors, the media and the Internet in creating model programs that will heighten visibility and give a stronger voice to the battle against galactosemia.

PGC is very proud and excited to be able to support such an important project and we are committed to continuing to support the work of Dr. Berry and his team at Children's. To launch the new center, Children's Hospital is seeking philanthropic support totaling at least \$1 million. Children's Galactosemia Center will have a profound impact on the health and well being of galactosemic patients and their families for years to come. Dr. Berry and other Children's leaders are now asking interested donors and friends to join them in making this one-of-a-kind center a reality. We hope that you will consider helping us reach the \$1 Million goal.

## PGC Announces 2008 Conference Location

The PGC Board of Directors has selected **Chicago, IL** as the location for the Summer 2008 Conference. Several factors lead to this unanimous decision including very reasonable quoted prices for rooms, food, and facilities, ease of air travel from all around the US and the world, nearby location of numerous family-friendly tourist attractions, and location of a PGC board member (Secretary Nish Rao) in the area to facilitate contact with vendors and service providers. PGC Sponsor, the Illinois Nut Company, is also located nearby and has promised to use industry contacts to secure favorable pricing for PGC on goods and services needed for a successful conference.

The board will work over the coming months to pick a hotel and lock down the dates. Then a conference committee will be formed under Nish's direction, to complete detailed planning.

Start making plans to visit the windy city in summer of 2008.

# Helpful Books About Premature Ovarian Failure

By Maureen Bell

I was eighteen years old when I was diagnosed with Premature Ovarian Failure (POF). When I was first diagnosed, I didn't think that much about it, because I was young, not dating, and not even thinking about marriage. Today, however, I am married, and premature ovarian failure is something I struggle with on a daily basis. If you are a parent of a young female with galactosemia, or are an adult galactosemic who has been diagnosed with POF, I strongly urge you to read the following books.

I first learned about the book "*Faces of POF—Learning and Living with Premature Ovarian Failure*" after attending the POF Conference in Virginia in 2004. The book illustrates the many different conditions that POF affects or has affected. It talked about how other women have dealt with POF and their stories. The viewpoints are diverse and it described a wide range of experiences of women with POF of all ages and backgrounds.

The following is a synopsis written by Nanette Santoro of the Albert Einstein College of Medicine:

*"This is a book about hope. It is a book about survival. You will not find helpless victims inside these pages. You will become acquainted with a series of strong women, the men who love them, and their families. You will learn how individuals cope successfully with the adversity of this very challenging condition".*

*Premature ovarian failure is, by all accounts, a devastating diagnosis. Peculiar symptoms often last for years before a correct diagnosis is made. Women with this disorder may not attribute their symptoms to menopause—why would they? Their health care providers also tend to misattribute symptomatology. Diagnostic testing is delayed, sometimes resulting in irreversible loss of bone density and premature osteoporosis. And the often irreversible and unexpected infertility that accompanies POF is frequently the most distressing blow.*

*How do women adapt to this unwelcome diagnosis? In my years of experience in providing medical care to women with POF, there are some common themes that emerge. One of the most powerful affirmations for women with this diagnosis is the knowledge that they are not alone and that they can help one another. The Internet has become a true information highway for women with POF. By uniting those with a relatively rare disorder, sharing of emotions as well as medical knowledge can provide much support. POFers can help each other find the doctors who will work with them, the hormones (or alternatives) that will keep their symptoms at bay and their bodies healthy, and the wherewithal to wait it out until some of the answers they need come to them.*

*In this way, POF victims can make the transition into POF survivors. In this book, you will find suggestions and hope that will help guide you or a loved one through this difficult diagnosis. There is no one, single answer for the infertility that so often accompanies POF: from adoption, to child-free living, to egg donation; the reader can try on each solution and see how it fits. Learning how to live with uncertainty is one of the central life lessons that POF forces women to acknowledge. Adaptation is an ongoing process that entails great personal growth, and this book will help to show you how to begin."*

The second book that I recommend is "*Infertility—Finding God's Peace in the Journey*" by Lois Flowers. This book answered questions about the who's, what's, and where's of infertility. But more importantly, it helped me "close the door" on POF by helping me to answer the question "why me?" This book helped me to understand that I am not alone in the journey, as many other women face the same situation as well.

Not only would I recommend this book for the woman with POF, but for her family as well. This book helps explain to others what they can do in terms of support, care, and overall understanding of the emotions involved with being infertile.

# **Generation G Program for Young Adult Galactosemics at the 2006 PGC Conference**

*By Linda Manis*

When PGC started over 20 years ago, the majority of the members were families whose galactosemic children ranged in age from newborn to ten-years-old. Now, those same children are in their twenties and early thirties – they are PGC's "1<sup>st</sup> generation of galactosemics" or **Generation Gs**. Thirteen GGs (as they are known), ranging in ages from 18 to 31, participated in the first-ever GG Program at the 2006 PGC Conference in Philadelphia, meeting in their own "Boardroom."

Galactosemics experience different levels of difficulties, and the GGs were no exception. The participants in this program ran the spectrum of difficulties, from mild to more serious. But that didn't stop this group from becoming tight-knit very quickly and forming a connection that only they can explain. A few GGs had NEVER met another person with galactosemia, while others had been to previous conferences.

Some galactosemics are shy, especially in new situations. But by the end of the first hour together, any anxieties had disappeared. Jim Svobodny (father of Brett – 18) and Joe Leggio (my new husband, stepfather of Adam Manis – 22; also the father of three grown daughters) ran the program, which Susan Fernstrom (mother of Holly – 28) and I organized and coordinated. Like most dads, Joe and Jim's enthusiasm and endless joking made the program extra special! PGCers who were in Atlanta or Reno with their kids might remember Jim. He's the one who took the kids to various offsite activities and more.

The GGs toured Philadelphia on "Ride the Ducks," on land and water in an amphibious vehicle. They took a trolley to a state-of-the-art bowling alley and spent the afternoon bowling. They had breakfast with Dr. Judy Fridovich-Keil, a biochemist who is doing research on galactosemia. All the GGs participated in her study by giving blood and filling out questionnaires.

## ***"No Parents Allowed"***

A few of the program activities were for the GGs only – "No Parents Allowed!" Sharon Howell, a 46-year-old woman with Galactosemia had a "Rap Session" with the GGs, where they discussed "What It's Like to Have Galactosemia." Laurie Bernstein, one of PGC's favorite registered dieticians met with the GGs to discuss their feelings and answer questions about the galactosemia diet. World renown galactosemia researchers and clinicians Dr. Louis Elsas and Dr. Gerald Berry had a private lunch with the GG group – Holly made sure that no one talked until the parents running the program left the room – telling us, "You parents have to learn how to let go."

## ***Other Stuff***

Sabrina Honeycutt (mother of an elementary age boy with galactosemia) and her fiancé Nick Elliott interviewed some of the GGs for a video fund raising project in South Carolina. The GGs played bingo (Matt Hynes – 31 – won \$20)! Then, an incredible interactive magician gave each GG a set of magic materials and taught them how to do magic tricks. On Friday night, the GGs and their families went out to dinner. The GGs sat together and for once were not alone ordering their selections without dairy. The parents sat at another table and compared their kids' experiences – crying, laughing, sharing stories, providing support, and learning from one another.

Over two days, the GGs made friendships that will last a lifetime. They were each given a disposable camera at the beginning of the conference to record their memories. At the end of the conference, each GG was given a binder with a photo of each GG and his or her contact information (e-mail addresses and birthdays too) along with a list of his or her favorite things, to use to stay in touch with one another until the next conference or get together. They have been e-mailing one another ever since.

## ***In Conclusion***

Almost everyone at the conference noticed the GG group, including teenagers who approached us to join the program, which was only open to those Galactosemics who were 18 or older. Those involved in the GG program and the PGC board agreed to expand the Generation G group to include any galactosemic who is 16 or older. This group will be completely independent of and separate from PGC, but PGC will still sponsor GG activities at conferences, in the

newsletter, and on the website. GG will be run entirely by volunteer parents of GGs. Susan Fernstrom, Rhonda Svobodny, and myself are willing to organize this new group. Our vision is a group of young adult Galactosemics getting together not only at the PGC conferences but once or twice a year – taking trips together (perhaps Disneyworld, a cruise, and/or having GG group get-togethers in the towns where various GGs live). If you are a GG young adult (age 16 to 35) and you and your family are interested in joining us, please contact one of us (contact info is below). If you are a parent who is interested in helping or planning, please let us know.

Words cannot describe the GGs' experience, but as one parent said, "This is the most wonderful time my child has ever had!"

### **Contact Information**

For more GG News please check out the Generation G link (which should be up in October) from the PGC Web site [www.galactosemia.org](http://www.galactosemia.org)

To join the group, either you or your parents can contact:

Linda Manis: [LMScript1@aol.com](mailto:LMScript1@aol.com) Phone: 561-862-0748 (Boca Raton, FL)  
Susan Fernstrom: [4willow@adelphia.net](mailto:4willow@adelphia.net) Phone: 508-498-0408 (Amesbury, MA)  
Rhonda Svobodny: [sherry7@aol.com](mailto:sherry7@aol.com) Phone: 916-635-4130 (Rancho Cordova, CA)

At the time of the writing of this article, I am trying to have a GG Section for the GGs to chat with one another on the Galactosemia Discussion Board at <http://www.galactosemics.com/>. Please check that site periodically to see when it becomes active.

## **Notes from Conference Presentations — Main Session**

### **Modifiers in Outcome in Galactosemia**

*By Bill Cozzo*

**Presenter:** Dr. Judy Fridovich-Keil, Ph.D.

Dr. Fridovich-Keil opened her talk by reminding us that people with galactosemia are individuals and, as such, experience the effects of the disorder in their own way. Of interest to the entire community of researchers, clinicians, families, and individuals with galactosemia is the question "why do some people experience long-term complications like learning or speech difficulties or ovarian dysfunction while others do not?"

Dr. Fridovich-Keil went on to explain that her research team at Emory University are 'digging deeper' by exploring biochemical and genetic factors beyond GALT and Gal-1-P. She is looking at factors such as glycolipids, the glycosylation state of FSH, and normal variations in other enzymes that metabolize galactose. She explained that understanding the factors that mediate long-term complications in galactosemia is the first step toward developing novel interventions to treat or even prevent them.

She indicated that studying these factors in real people with galactosemia, both those experiencing complications and those who are not, is the key to continuing progress in the development of treatments. To that end, she invited all galactosemics present at the conference (or their parents), and those reading about this now, to contact her to arrange to submit a blood sample and fill out a questionnaire. Nearly 100 people have signed up to participate thus far, and that is a tremendous start. But more are needed. Please consider helping this very worthwhile research the next time your child needs to have a blood test. See the PGC website for details on contacting Dr. Fridovich-Keil for detailed instructions.

All of Dr. Fridovich-Keil's slides featured snapshots of smiling people of all ages. There were pictures of children at play, pictures of families celebrating graduations, weddings, and enjoying holidays. Most of these charming faces were of galactosemics, but many were not. Her warmth and compassionate approach gave the audience a deep sense of Dr. Fridovich-Keil's commitment to helping real families dealing with their real life challenges and celebrating their real life victories. We look forward to hearing the results of her investigation in the future.

# Ask The Experts Panel

By *Therese Cozzo and Kristine Lydon*

## **Panel Participants:**

**Dr. Berry, Dr. Elsas, Dr. Nelson, Dr. Potter, William Bell RPH MBA, Laurie Bernstein MS RD FADA**

### **Question #1**

Do males who have galactosemia experience any problems with testosterone levels or infertility?

#### **Answer:**

Dr. Berry explained that there are no known abnormalities for males who have galactosemia with regard to testosterone function or reproduction. He went on to explain that there are very few studies in this area. He referenced a Swiss study that determined there was no difference in male characteristics or testosterone between males who have galactosemia and those who do not. Dr. Berry pointed out that there are no studies on sperm production or maturation. He further explained that there are few reported cases of male galactosemics fathering children.

Dr. Elsas added to the topic by explaining that females develop their eggs in utero (ovaries exposed over time) while the in males, gametes are not made in utero.

### **Question #2**

Which is the best indicator of outcome: Gal-1-P, breath test, or galactose?

#### **Answer:**

Dr. Elsas explained that Gal-1-P, breath test and galactitol are all predictors of outcome, but said that the best predictor for “dyspraxic speech” was the breath test. He said that the best predictor of cognitive outcome was unknown. Dr. Elsas stated that we need a center of excellence and enough patients to study these factors

### **Question #3**

How often should patients get their blood tested for Gal-1-p levels after age 18?

#### **Answer:**

Dr. Berry shared that his office recommends that patients over the age of 18 be tested once a year for gal-1-P and galactitol. He further explained that the need to test blood for older patients is not well established, and suggested that these tests may be eliminated in the future as most patients Gal-1-P levels stay relatively constant after age 10.

### **Question #4**

The panel was asked to address if a “relaxed diet” for patients over 3-5 years of age may lead to improved outcome?

#### **Answer:**

Laurie Bernstein shared that based on data from patients, her clinic allows all fruits and vegetables. They are not relaxing the diet more than this at this time, but are continuously watching/listening to researchers.

Dr. Berry shared that there is some research outside the United States that shows some patients may do better on a more relaxed diet, but there is no evidence in the United States that suggests that liberalizing the diet is best (in this usage, “very liberal” is taken to mean lactose ingestion). Dr. Berry suggests that this area is one where we need to study. He finished by stating that he is NOT advocating a very liberal diet at this time.

Dr. Berry discussed research by a German group - patient consumed 1300 mg galactose/day - did not result in sustained increase in gal-1-p or galactitol.

Dr. Elsas - suggested that alternate pathways might be involved - just because gal-1-p did not increase, does not necessarily mean that cells are protected.

Laurie addressed parents' concerns that diet infractions cause ovarian failure. Laurie suggested that these things are *predetermined* and not based on diet infractions. She recommended to parents that they be the advocates for their children by talking with their doctors and making informed decisions. *Make decisions based on your own child - is it the right choice for him/her?*

**Question #5**

For Dr. Nelson: How long should someone be on hormone replacement therapy?

**Answer:**

Dr. Nelson recommended that hormone replacement therapy continue to the typical age for menopause (age 50). He also said that an individual could continue with therapy until age 55 or stop at age 45 (based on individual variations).

Dr. Nelson recommends an estradiol patch because he feels it provides the most natural delivery - into the veins and gets more estrogen into the blood. He stated that the estrogen dosage is higher in oral estrogens, although, if a patient can't use the patch, oral estrogens are ok.

**Question #6**

For Dr. Potter - Is it helpful from a speech and language development perspective for our children to sing songs? Is it helpful or harmful to try to teach our children to be bilingual?

**Answer:**

Dr. Potter: Singing songs is helpful because it involves playing with words and songs, but it probably does not serve to make actual speech better. A bilingual environment slows down language by several months, but will not hinder language acquisition. She feels it is ok to raise a child to be bilingual, but recommends no more than 2 languages. She also recommends teaching the languages with very low stress for the children.

**Question #7**

Question for Bill Bell: Recommendations on traveling and refilling prescriptions away from U.S.?

**Answer:**

Bill stated that ingredients and regulations for medicines might be different from country to country. Bill advised that people fill prescriptions at home and bring plenty along when traveling outside the U.S. He also recommended that prescriptions be placed in carry-on luggage.

**Question #8**

Panel asked if stem cell research could help galactosemia and if partial liver transplants could help to cure neurological problems seen in galactosemia?

**Answer:**

Dr. Elsas replied, "No." to both questions. He further explained that transplantation is very invasive, and there are problems with immunodeficiency.

Dr. Berry stated that there is one known patient who had galactosemia and another genetic disease who had a transplant. It is unclear if this transplant affected the patient's Gal-1-p levels. Dr. Berry thought that partial liver transplant was not an effective way to go in treating galactosemia. He thought that the research being conducted in other disorders might serve to help us understand galactosemia better. Dr. Berry stated that it is important to know when toxicity occurs, that is, pre-natal or post-natal. If it occurs prenatal, we would need to develop a therapy *in-utero*. Dr. Berry stated that no one is entertaining stem cell research in this area.

Dr. Berry suggested that we need to understand basic physiology and explained that we don't know the long term effects of transplantation. If endogenous production of galactose is the problem then liver transplantation may not help.

**Question #9**

How much galactose does the body actually produce, and what is the relationship of this endogenous production to food (galactose) intake?

**Answer:**

Dr. Berry stated that a method had been developed to measure the endogenous production of galactose, but that this production is an apparent appearance of production rate (i.e., an approximation). He estimated endogenous production of galactose to be:

*Infants and Children:*     1.34 mg/kg/h  
*For Adults:*                 0.56 mg/kg/h

## Ask The Experts Panel (continued)

Dr. Berry indicated that galactose production might be related to growth.

Dr. Berry referenced that Dr. Segal saw no increase in gal-1-p for two adult patients with galactosemia, but he recommends further study in this area to find the safest approach.

### Question #10

What is known about the percentage of woman with galactosemia having children?

#### Answer:

Dr. Berry referenced early reports concerning African American woman with the S135L mutation (less severe mutation) having children. He suggested that among Caucasian women with galactosemia, less than 10 Caucasian women with galactosemia have children.

Dr. Elsas also shared that originally, data suggested that 85% of woman with galactosemia experience premature ovarian failure, and only 15% would be expected to have children. He also stated that all of this data came before higher sensitivity to newborn screening, and suggested that perhaps earlier detection of galactosemia may lead to an increase in birth rate.

### Question #11

Is it safe for a woman with galactosemia to breastfeed ? (Does lactation increase galactose levels in the mother ?)

#### Answer:

Dr. Berry responded that there are some studies that indicate that lactation does affect gal-1-p levels. Lactation may cause lactose to be produced, and result in galactose levels to rise. The clinical effects of this are not known. When asked by Dr. Elsas if endogenous production increases in a woman during pregnancy or in the postpartum state, Dr. Berry said that endogenous production may increase, but this has not been studied.

### Question #12

Are there reports of disfluency (i.e. stuttering) in children who have galactosemia?

#### Answer:

Dr. Potter responded stating that she only tested 15 children who have galactosemia in her recent study and only one of the 15 families mentioned stuttering as an issue. She doubts a connection between galactosemia and disfluency.

### Question #13

Are soy products, soy flour, tofu, and etc. acceptable in the galactosemic diet?

#### Answer:

Laurie stated that her clinic has not seen a rise in Gal-1-P when patients were given soy products. Her clinic allows *all* soy products. Dr. Berry and Dr. Elsas both agreed that soy products were ok.

Laurie added that she does not think that isoflavins (found in soy) are involved in premature ovarian failure

### Question #14

Is there a "standard of care" for patients who have galactosemia (i.e., how often to get gal-1-p levels or galactitol levels checked, when to see an Ob/Gyn, etc.?)

#### Answer:

Dr. Berry recommended that a multi-center study would be best to get a group of centers to do a prospective study in order to study gal-1-p and galactitol levels and correlate these levels to clinical parameters. Dr. Berry said that with this information, we could decide what information is most meaningful. Dr. Berry stated that, ideally, we want to do the minimal number of testing and keep it safe. He recommends Gal-1-p testing every 3 months for children birth to age 1 year, Every six months the second year, then every year after that.

Dr. Berry found that galactitol did not provide any better information than gal-1p, but helps in newborn screening.

**Question #15**

When should we do bone scans?

**Answer:**

Laurie explained that the problem with bone scans/dexa scans is that they give a "z score" that is based on *adult* scores, and therefore, are not really relevant to children.

Dr. Elsas suggested a baseline scan at 9-10 years of age.

**Question #16**

When should we bring our daughter to see a gynecologist?

**Answer:**

Dr. Nelson recommended that girls should see a gynecologist or pediatric endocrinologist if they have no signs of puberty by age 14, or if they do not get their period by age 16.

Dr. Elsas stated that this decision should be made on *individual* basis suggested that this answer may vary for patients who are homozygous vs. heterozygous.

Dr. Berry suggested that patients act in a preemptive way and he recommended that patients see an endocrinologist sooner because of the tremendous problems. Dr. Berry suggests that the rate of POF in patients who have the Q188R/Q188R mutation is 100%.

Dr. Elsas adds that screening too early (e.g. 3-5 yrs of age) for FSH levels can lead to false positive results.

### *With deepest sympathy ....*

PGC extends our condolences to the Baigent family from Canada for the recent loss of their loved one, Taylor Baigent. Taylor supplied PGC with our lapel pins, unique for each conference, for many years. His wife and children have also been involved with our organization for a very long time. Taylor was a wonderful man and will always hold a special place in the memory of PGC.

# Speech Disorders and Galactosemia

By Nancy Potter, Ph.D., CCC-SLP

An estimated 60% of children with galactosemia have speech disorders. At the 2006 Parents of Galactosemic Children Conference, I had the opportunity to speak with parents of children with galactosemia who have speech disorders and I am grateful for this opportunity to address their most frequent questions.

## **What is a speech disorder? (Actually, this question was not asked but the terminology is necessary to address the following questions.)**

A speech disorder is any deviation of speech that is outside of the acceptable variation for a given environment.

These deviations include the areas of:

- 1) *Audibility* — speech is too loud or too soft
- 2) *Intelligibility* — speech is difficult for everyday listeners to understand
- 3) *Specific sound production errors* which include:
  - Omissions (example: ha instead of hot)
  - Substitutions (example: fumb instead of thumb)
  - Distortions (the sound can be identified but is not produced accurately)
  - Additions (extra sounds are added to a word or between words)
- 4) *Prosody* — different rhythm or stress
- 5) *Pitch* — too high or too low
- 6) *Voicing* — over-voicing or under-voicing (whispering)
- 7) *Interruptions* in voice, sound production, or rhythm

## **What are the different types of speech disorders?**

Speech disorders are typically differentiated into two categories of articulation or phonological process errors and speech motor impairments. An articulation disorder is a problem in producing one or more specific speech sounds. The most common articulation errors are the sounds /r/, /l/, and /s/. A phonological process error is a pattern of simplifying difficult sound productions usually through omission or substitution. Articulation and phonological process errors are the most common types of speech errors and the easiest to correct.

Speech motor impairments (also termed motor speech disorders) include apraxia of speech and dysarthria. There are many different terms used to refer to apraxia of speech in children including developmental verbal apraxia, developmental apraxia of speech, developmental verbal dyspraxia, and childhood apraxia of speech. The American Speech and Hearing Association (ASHA) recommends the use of the term childhood apraxia of speech (CAS) because the term 'developmental' implies that the child may outgrow the disorder; therefore some third party payers (insurance companies) have denied reimbursement for speech-language services due to the inclusion of the term developmental in the diagnosis. The term dyspraxia (reduced function) is used in Europe and Australia while apraxia (absence of function) is used more frequently in the U.S. Apraxia involves a disruption in the planning and programming of speech. The child with CAS knows what he or she wants to say, but has difficulty planning and programming the mouth, lips, tongue, jaw and voice movements required to successfully produce intelligible words and sentences.

Childhood dysarthria is a disruption in the execution of movement used to produce speech due to central (brain and spinal cord) or peripheral (spinal and cranial nerves) dysfunction. Speech characteristics may include hypernasality, imprecise consonants, vowel distortions, and rate control problems.

## **What types of speech disorders are typical of children with galactosemia?**

Apraxia is the most common diagnosis given to children with galactosemia and speech disorders. Our recent study (15 children with classic galactosemia and speech sound disorders) found that children with galactosemia have a mixed form of apraxia and dysarthria.

## **My child does not qualify for services based on his or her performance on articulation tests, but most people have difficulty understanding him or her.**

Children (and adults) with speech motor impairments frequently have more speech errors as their sentence length increases. In addition, prosody errors become more apparent with increasing sentence length. A good measure of intelligibility is to calculate the percent of consonants correct and the percent of vowels correct in recorded conversation. When the percents are below 85-90% correct, it becomes difficult to understand a person's speech.

**Our school district denied services to our child because of a low or low-normal IQ. Is this a factor in the severity of their speech disorder?**

Our study found that the severity of the speech disorder was not related to IQ. Children with higher and lower IQs had similar speech disorders. A low IQ score should not limit speech services for children with galactosemia.

**How many speech therapy sessions will be required to improve my child's speech?**

Thomas Campbell at Children's Hospital of Pittsburg found that children with severe phonological disorders required an average of 29 treatment session to improve from less than 50% intelligible speech to 75% intelligible speech while children with apraxia required and average of 151 sessions to achieve similar improvement.

**Will oral motor exercises improve the speech of children with speech motor impairments?**

Probably not. There is no peer-reviewed evidence that oral motor exercises improve speech. While oral exercises and speech both use the articulators, they do not stimulate the same areas of the brain. I recommend focusing on speech rather than exercises during speech therapy sessions.

**Our speech language pathologist is willing to work with our child but has not had experience with speech motor impairments. Are there any references that you recommend?**

I found the book Clinical Management of Motor Speech Disorders in Children by Anthony Caruso and Edythe Strand (1999) helpful in structuring effective speech therapy sessions.

Nancy Potter, Ph.D., has worked as a speech language pathologist for more than 20 years, specializing in childhood speech motor impairments. She is an assistant professor at Washington State University-Spokane and recently completed a study with Lawrence D. Shriberg, Ph.D., University of Wisconsin-Madison, examining the speech, language, and motor characteristics of children with galactosemia.

## ***Recycle Empty Ink Jet Cartridges / Retired Cell Phones and Help PGC Raise Funds***

PGC has joined forces with AAA Environmental, Inc. to institute a *new and exciting recycling program* that will remove unwanted plastics from the landfills and can provide a positive cash flow for the Parents of Galactosemic Children, Inc.

AAA Environmental, Inc. will supply us with NO COST postage paid recycling envelopes to send in our used printer cartridges and retired cell phones. We have recycling envelopes for you to use or give to family, friends, neighbors, etc., anywhere in the United States.

For more information or to obtain envelopes, please contact  
Michelle Fowler at [michellefowler1@bellsouth.net](mailto:michellefowler1@bellsouth.net).

Notes from Conference Presentations — Main Session  
**Speech, Language, and Motor Characteristics of Children with Galactosemia**  
*From notes summarized by Nish Rao*

**Presenter:** Nancy L. Potter, Ph.D., CCC-SLP

The subject of this talk was speech and sound disorders in galactosemic children. Dr. Potter talked about past and current research studies and the direction future research projects will take.

At the beginning of this presentation, the audience was asked to participate in some examples of speech therapy exercises. First was a three-step oral motor exercise where Dr. Potter had the audience touch their lips together, then touch the tip of their tongue to the roof of their mouth, then touch the underside of their tongue to the roof of their mouth, all in rapid succession. Next, came a quasi-speech task. The audience was asked to say “pa-ta-ka” five times in a row, starting off slowly, then building up to very quickly. The final example was a speech task of 15 syllables (*I pledge allegiance to the flag of the United States of ...*) At the end of this exercise, Dr. Potter emphasized that just because a speech-like task is used in evaluation, it does not mean the task should be worked on in therapy. Therapy should focus on aiding/enabling a better result from the oral motor exercise.

Two types of speech/sound disorders were mentioned.

- Phonological or articulation disorders where specific sounds or groups of sounds are produced incorrectly at least 40% of the time
- Speech Motor Impairments
  - Apraxia - Difficulty in planning and programming speech
  - Dysarthria - Difficulty in producing intelligible speech due to a neurological disorder

It was mentioned that there are different diagnostic criteria for apraxia in children and in adults across the country and around the world. Dr. Potter recounted how the state-of-the-art among researchers in this field is still undecided. A recent meeting of top professionals in the field could not come to a common agreement on exactly what the definition of apraxia of speech should be. So research continues.

The question is, can an adult still have the disorder known as “childhood apraxia of speech”? The approach has been to examine populations diagnosed with apraxia and redefine criteria for speech motor impairments

Previous studies have reported that:

- 50-60% of children with galactosemia have speech disorders
  - Typically diagnosed as “apraxia” (Nelson et al., 1991, 1995, Webb et al., 2003)
- 90% of children with galactosemia and “speech” disorders also have “language” disorders (Waggoner et al. 1990)
- 18% of children with galactosemia have motor/coordination disorders (Waggoner et al. 1990)
- Motor disorders frequently co-occur in children with speech motor impairments (Dewey et. al., 1993)

The purpose of Dr. Potter’s investigation was to describe the specific speech, language, and motor characteristics of children who have galactosemia and speech disorders. The research questions asked were:

**Q1.** What are the characteristics of the persistent speech, language, and motor impairments in children with galactosemia?

**Q2.** What are the relationships among speech, language, motor, and cognition in children with galactosemia?

Participants in this research group were 15 children with classic galactosemia and either current, or a history of, speech sound disorders. The children who participated in the study were between ages 4 and 14. They were recruited through PGC's mailing list and website. These tests were conducted at their homes.

Standardized test methods were used for this research, including, *Goldman-Fristoe Test of Articulation-2 (GFTA)*, *Oral and Written Language Scale (OWLS)*, *Kaufman Brief Intelligence Test (K-BIT)*, and the *Movement Assessment Battery for Children (MABC)*. Conversational language samples were analyzed with both SALT (Systematic Analysis of Language Transcripts) and PEPPER (using phonetic transcription). Additionally, the children were given challenging word tasks. Each child's case history was gathered from the parent, and each child was given a oral structure and function exam.

Dr. Potter shared that there were some consistent speech characteristics found with the galactosemic children she met. These included persistent speech sound errors which should be gone by age 9 in a non-affected child, and differences in timing and stress of speech, known as *prosody*. Additionally, she noted that speech errors increased with longer words and phrases in the children she observed. Interestingly, the severity of speech impairment was not related to IQ of the child. These observations are consistent with a diagnosis that indicates speech motor impairments such as mixed Apraxia and Dysarthria.

Half of the children tested had hoarse or breathy voice, or occasional aphonia (periods of no voice or a whisper.)

Some language characteristics that these children showed included standardized language test results (OWLS) that were 36% below the 'normal' range expected and conversational language results (SALT) that were 64% below the 'normal' range expected. These are consistent with speech motor impairment.

Analysis revealed the presence of significant deficits, including more omissions (words and bound morphemes) as utterance length increased, smaller vocabulary (fewer # of root words), slower speech (fewer words/minute), and shorter phrase and sentences length.

Dr. Potter's work showed that some aspects of speech / language difficulties were related to IQ, wherein those with lower IQ had shorter phrase and sentence length, and lower standardized test scores for language (OWLS) and motor function (MABC). But there were other aspects of speech / language difficulties that occurred without a relationship to IQ. These were articulation, intelligibility, speech rate, word and morpheme omissions, and vocabulary.

Dr. Potter's impressions were that these children (and parents) are hard working, cooperative, and very perceptive. They have many interests and hobbies. These children are good conversationalists and are not shy, contrary to the reporting of Fisher et al. 1980 regarding children with galactosemia.

Dr. Potter indicated that children with galactosemia and speech disorders can have persistent speech sound errors consistent with a speech motor impairment (e.g. mixed apraxia and dysarthria), show increasing speech AND language errors as length and complexity of utterance increases, and continue to use slow and short utterances into the adolescent years due to breakdowns in speech and language with increasing utterance length. These children might also need to be tested for speech and language impairments using connected speech in order to evaluate the frequent and unusual errors. Dr. Potter shared that children who are evaluated using only standardized tests may be denied services even when significant impairments exist.

Dr. Potter concluded by indicating that there are ongoing metabolic studies with Emory University that hope to examine the differences in outcome measures related to metabolic efficiency due to genotype. The focus will also be to examine differences in white matter formation in the brain using neuroimaging. Dr. Potter invited the audience to become involved in the research by contacting her via email: [nlpotter@wsu.edu](mailto:nlpotter@wsu.edu)

## Notes from Conference Presentations — Main Session Rx for Success

*From notes summarized by Nish Rao*

**Presenter:** William F. Bell, Jr. R.PH., MBA  
Senior Clinical Pharmacy Specialist  
Gould and Lamb, LLC.

At the beginning of his presentation, Bill talked about choosing a pharmacy/pharmacist that is best for you and your Galactosemic child. Ideally, you should find a pharmacist who will be able to learn and understand about galactosemia, and take the time to answer your questions. Further, they should be available to relay important information in an emergency and have a good rapport with your child's medical team (pediatrician, geneticist, etc). Hopefully they will be honest enough to say, "I don't know" and committed enough to say "I will find out" when necessary.

Next, Bill talked about Lactose Free Medications. He indicated that ingredients change all the time in prescription and over-the-counter medications, much like food items and cautioned not to assume that medications are lactose-free just because "they use to be". He named some current medications that are currently lactose free as a pointer on where to start if the need arises. These include:

Amoxil, Amoxicillin, Biaxin  
Zithromax  
Tylenol OTC, Benadryl OTC,  
Robitussin  
Phenergan Cough Syrups  
Flavor -RX  
Claritin

Bill continued by addressing the topic of hormone replacement medications. He shared that Demulen has started using lactose and that other acceptable hormone replacements are Ortho-Evra Patch, Vivelle Dot, Transdermal Estrogen Replacements. He then discussed ADHD / ADD medications and cautioned that most, if not all, contain lactose. He advised parents to collaborate with their child's physician before use. The presentation then became a discussion as the opinion emerged stating that small amounts of lactose in necessary meds might not be harmful, especially compared with the benefit they provide and the much larger amount of endogenously produced galactose.

When Bill continued his talk, he covered the topic of calcium supplementation and cautioned that multi-vitamins are not enough. The Recommended Daily intake of calcium is 1000 –1200 mg per day for adults and 800 mg per day for children. He also advised that diet and weight-bearing exercise together can help in fighting Osteopenia / Osteoporosis.

He concluded by offering some recommendations on resources and information available to help when researching medications. These Include:

NeedyMeds.com  
Google  
<http://www.hshsl.umaryland.edu/consumer/sites/druginfo.html#drug>  
Your Pharmacist!

## New Additions to PGC's leadership team

The Board of Directors of PGC is happy to announce the addition of Paul Fowler, Nishkala Rao, and Bill Cozzo to the Board of Directors. Paul will serve as the Treasurer and be responsible for keeping track of PGC's savings, donations and expenditures, having the books audited annually, and preparing financial statements twice per year for publication. Nish will serve as secretary, maintaining the minutes of Board meetings, as well as other official correspondence and records of the organization. Bill will serve as Director-at-large and handle tasks of Outreach Committee chair, PGC's website coordinator, and Newsletter editor.

# The Galactosemic Diet — Updates Since the Conference

Laurie Bernstein MS RD FADA

The PGC Conference was a wonderful venue for all of us to meet. In a relatively short period of time I had an opportunity to share a lot of information. I will review what appears to have caused some confusion, and answer some questions that have been posed to me since we have all returned home.

## A Quick Review of Dietary Sugars

A monosaccharide (having 1 Hexose unit) is a simple sugar containing six carbons. Examples include the sugars glucose, fructose, and galactose. Disaccharides (2- Hexose units) include Sucrose (which breaks down to glucose and fructose), Lactose (which breaks down to glucose and galactose), and Maltose (which breaks down to glucose and glucose). The Tri-saccharides (which have 3-Hexoses units) are Raffinose and Starchyose. The tri-saccharides are found in beans and legumes. They are considered bound galactose and cannot be digested by our small intestine and end up being metabolized and expelled from the large intestine.

## Dietary Sugars in the Galactosemic Diet

Humans do not present with alpha amylase enzymes needed to breakdown and release galactose from Raffinose and Starchyose.

Please note that the Denver clinic **does** restrict Garbanzo beans, Split pea (green and yellow) Lima Beans, Lentils, Pinto, and Kidney beans. A few families at the conference shared they have never restricted these products and gal-1-p levels remain within treatment range.

It is fair to say that the Denver clinic **does not** restrict any fruits or vegetables in the diet of galactosemia patients. We **do not** restrict soy products. We do recognize that some tomato products are higher than some of the beans (i.e.: pinto) that we do restrict. We have not seen a correlation with gal-1-p levels and fruit and/or vegetable intake. In fact, our observation has been as the child gets older and he ingests more of these products his levels often drop. This observation might be more closely related to age and development on a cellular level than to actual intake. A review of our legumes restriction is ongoing.

## Sodium Caseinate:

Sodium caseinate is the sodium salt of casein, a milk protein. This question came with regard to Coffee-Mate Liquid and Powder.

Nestles responds with the following:

*"COFFEE-MATE Liquid and Powder products are non-dairy and are Kosher according to the Orthodox Union (as indicated by the "O.U." symbol)."*

*As a courtesy (emphasis added), we place a "D" next to the kosher symbol (O.U.) to alert those who adhere to strict religious practices. COFFEE-MATE contains an ingredient called sodium caseinate, which is a milk derivative, though it's classified as a non-dairy product. COFFEE-MATE Latte Creations and products contain both milk and lactose."*

*How is this so?* When sodium caseinate is processed, it is so materially altered that both dairy scientists and government regulators no longer regard it as a true dairy substance. This is why sodium caseinate can be an ingredient in non-dairy products, according to FDA, regulation 21 CFR 101.4 (d). Sodium caseinate is also not a source of lactose.

*So, where does this leave the families?* On one hand we agree that we will allow the foods made on machinery that has produced products with milk, therefore, we disregard the D. Is the processing of sodium caseinate sufficient to disregard the D and allow this ingredient in the galactosemic diet? I say yes, but please check with your metabolic nutritionist.

*continued ...*

## The Question of Cheese?

The Schaerdinger Dairy Association in Austria uses special processing in a salt bath that, combined with time, results in undetectable levels of lactose and galactose in ripe **Emmentaler** cheese. This is also true for **Gruyere and Tilsiter** cheeses. A sample of the process is detailed below; this may shed some light on the procedure. What it means in layman's terms is that the lactic acid process stops when all the lactose is gone. Our patients have used these cheeses from Austria and Switzerland without any increase in levels. I found this information about the manufacture of these cheeses.

**Flavor:** typical flavor developed by red and yellow smear producing bacteria during ripening for at least 4 weeks

**Ready for consumption:** after at least five weeks

### METHOD OF MANUFACTURE

**METHOD OF COAGULATION:** rennet or any other suitable coagulating enzymes, lactic acid starter

**HEAT TREATMENT OF THE COAGULUM:** scalding after cutting the coagulum

**FERMENTATION PROCEDURES:** lactic acid fermentation

**MATURATION PROCEDURE:** ripening at 12°C – 16°C

**OTHER PRINCIPAL CHARACTERISTICS:** salted in brine

The next question was regarding **aged cheeses**. I cannot answer this question. I have researched this question a fair amount since the conference, and I do not feel comfortable with the data I found. A conglomerate of information from many sites is listed below:

*“Here is a sure way to tell that a cheese is fermented. Fermented cheese has holes in it, because the microorganism produces gas during the fermentation process. In firm cheeses, like aged cheddar and Parmesan, the pressure of forming the cheese has compressed the hole, but if you look closely a kind of layered flakiness will reveal there were gas spaces before pressure was applied, while the high lactose processed cheese will be velvety smooth. The exceptions to this rule are the soft runny cheeses like Brie and Camembert, which are smooth but low in lactose.”*

“Low in lactose” - What does this mean?

*“Cottage cheese, theoretically, should be low in lactose (??) since its traditional preparation is heating fermented milk until the curds clump. However most cottage cheeses sold in North America today are “creamed”, meaning that cream, milk or milk solids are added to the cottage cheese. Usually among the plethora of adulterated cottage cheese products, one can find cultured dry cottage cheese (often packaged in a plastic bag) that is quite safe to use as an ingredient in a lactose free diet. “*

This information does not offer me a lot in guiding you. I do have some phone calls pending about the methods used for the various aged cheeses in question. As I continue to explore this, I will share my findings with the PGC. I hope the question of the Emmentaler, Gruyere and Tilsiter has been answered.

The questions around diet continue, I urge you to speak with your metabolic nutritionists.

Don't forget AllergyKids.com

Let them know you are with PGC – they have special products that will interest you.

I hope to see you all again soon.

Notes from Conference Presentations — Main Session  
**Imaging Studies: PET Scan and Functional MRI**  
*summarized by Bill Bell*

**Presenter:** Dr. Andrew Newberg, M.D.

Dr. Andrew Newberg from the University of Pennsylvania spoke to the general audience concerning functional neuroimaging and the implications for the galactosemic patient. Dr. Newberg spoke that the PET scan needs the most focus, since it can give us the most information about brain activity and function. He concluded that a galactosemic might show decreased activities in a PET scan. Dr. Newberg also mentioned that there is more work to be done in this area and that we have a long way to go before the routine use of neuroimaging of this type is widely recommended.

The functional MRI, or FMRI, is also an important tool in which to understand and decipher brain activity in galactosemics. Dr. Newberg also pointed to four main topics for future application of this type of imaging. These topics are: diagnosis based on metabolism, blood flow, or neurotransmitters, activation studies to assess effects of the disease on the brain, following the course of the disease, and following the effect of interventions.

Notes from Conference Presentations — Breakout Session  
**Premature Ovarian Failure**  
*summarized by Bill Bell*

**Presenter:** Dr. Lawrence Nelson, M.D.

Dr. Lawrence Nelson of the NIH in Bethesda, Md. addressed a breakout session for the 2006 Parents of Galactosemic Children Conference. The breakout session was titled "Premature Ovarian Failure (POF)." During this breakout session, Dr. Nelson discussed that emotional health plays a big part in helping the patient after the diagnosis of POF. Dr. Berry also was in attendance and offered insight on the prevalence and background of POF in the galactosemic female.

Dr. Nelson and Dr. Berry spoke about many issues in dealing with the diagnosis and long-range outlook in females diagnosed with POF. Issues ranged from "when to tell a female she may become infertile" to the importance of parents and patients getting their emotional "health" in check after the diagnosis is made.

Since the conference, both doctors have been in communication regarding the linkage of POF and galactosemia. A future meeting is planned for 2007 where the hope is to bring together women with POF from all aspects of life. The purpose of this meeting will be to discuss better ways to deal with POF emotionally and what the best treatment for patients is. More information will follow at a later date.

*With the end of 2006 right around the corner, please remember to keep PGC in your year-end plans for charitable contributions. All donations to PGC are fully tax deductible. Send your donations to :*

**Parents of Galactosemic Children, Inc  
P.O. Box 2401  
Mandeville, LA 70470-2401**

from Conference Presentations — Breakout Session  
**Speech: Beginning Talkers**  
*From notes provided by Kellie Wilcox*

**Presenter:** Dr Nancy Potter

Dr. Potter's talk covered treatment of childhood motor speech impairments and things parents should know and do as they begin to teach their child to speak. This material can assist the parent in being a better-informed 'consumer' of speech therapy as well help the parent teach their child to speak.

Dr. Potter shared that a good speech therapy plan should identify the specific problem(s) the child is having, a specific goal to address the problem(s), and a plan to reach the goal(s).

Dr. Potter indicated that the activities in her presentation could be used with a two-year-old child as long as the child can maintain attention, make and keep eye contact, and has a willingness to imitate.

The 'hands on' part of the session focused on an Integral Stimulation Approach.

Dr. Potter demonstrated ways that parents could show the child various mouth movements that are related to speech and have the child imitate it. Then she suggested a way to teach a child to say a simple word by starting with the end sound first, then adding the beginning sound to the ending sound.

Example: Teaching the child to say the word "fit"

- start with making the end sound, "-it" and have the child repeat it several times
- then add the starting sound "f"

Some of Dr. Potter's other exercises were:

Exercise:

*Point to simple body parts (e.g. nose, ears, head, etc) and say the name  
Then let the child repeat*

Exercise:

*Say an animal's name and make the animal sound (e.g. sheep – Baa Baa)  
Then let the child imitate the animal sound  
Then change the sound (Bee Bee, Boo Boo, Bye Bye)  
Repeat, Repeat, Repeat*

Exercise:

*Pick 5-10 words with the same sound (e.g. P sound)  
cuP, moP, hoP, Play, Pop  
Go over words and the sounds, saying the "P sound" over and over (e.g. P P P P P)*

Dr. Potter suggested that the parent work on these for 6-8 weeks at 10-15 min. a day or twice a day, then work on another sound, or a sound that builds on the first sound (e.g. first learn the P sound, then learn the P sound combined with a vowel sound, such as Pa)

Dr. Potter indicated that at 24 months old, a child should have 50 words and some two-word phrases. A late talker will not be doing this. About 25% of kids will not be doing this and will need help.

*continued next page*

If you would like to receive e-mails from PGC concerning such things as food item recalls, items of interest regarding galactosemia, and upcoming PGC events, please forward your name and email address to

**emaillist@galactosemia.org**

Please put "PGC Mailing List Addition" in the subject line of the email

## Speech: Beginning Talkers (continued)

Dr. Potter then shared some short sentence activities:

### Exercise:

Two word sentence where one word stays the same and the other word changes  
(e.g. my *blank*)

*my doll*

*my cup*

*my dog*

### Exercise

Gradually add one word at a time

*my doll house*

*my cup milk*

60% of galactosemics will have speech problems. Children have speech problems and/or language problems. Parents should be able to tell a speech problem by age 4.

In describing ways to work on speech with a child, Dr. Potter suggested working on 5-6 words at a time for severe problems and 8-10 words at a time for moderate problems. Work on enough words to master what you are working on so the child feels they have accomplished something. After the child masters some sounds and words, begin a random learning process to add new words.

## Notes from Conference Presentations — Breakout Session

### **Diet: Birth to Five Years**

From notes gathered by Katrin Muir

**Presenter:** Laurie Bernstein MS RD FADA

Laurie started off by retelling the well-known “Welcome to Holland” story (see PGC’s Website to read it). The story’s comforting message is that once we get use to the situation of having a child that might face challenges, we can learn to “enjoy Holland” just as much as we would’ve “enjoyed Italy”.

Laurie offered that nutritionists are here to help the parents and the children for life and emphasized that it is important for parents to have a good relationship with their child’s nutritionist. She sympathized that working with a nutritionist might be very emotional for some new parents. These parents might feel as though the nutritionist is, in effect, taking away one of the most basic things that parents do for their child - feeding them. Parents who accept the help of a nutritionist as a partner in caring for their child will feel the most comfortable.

Laurie then covered some basic background information in order to fill in any gaps in understanding that we may have. She gave a brief overview of the biochemistry of nutrition to describe the basis of the disorder. Then she described a bit about the genetics of galactosemia. It is an autosomal recessive disorder, which means that both parents contribute a defective gene that leads to galactosemia and no one is “to blame” for causing it. Laurie continued by telling us that “genetics has no memory” and that the risk of having another child with galactosemia is 25% with each pregnancy. She finished this section by talking a little bit about the outcome for galactosemics. She indicated that with galactosemia, we cannot know at the time of birth and diagnosis what the outcome will be for that child. We can prevent cataracts and liver disease by following the diet, but some of the complications may be already determined before birth.

Laurie shared about how she talked with fifteen adolescents with classic galactosemia. They each believed (wrongly!) that they would die (!) if they ate something that is forbidden. Laurie emphasized that parents should not

tell their children that certain foods “will make you sick.” If parents tell that to their child, and the child eats the food (either accidentally or intentionally) but *doesn't* get sick, the parent's credibility is ruined.

Laurie gave a wonderful description of parents' emotional responses to the diagnosis of a chronic metabolic disease like galactosemia. These responses are natural, but the parent must be sure that they don't “get stuck” in any of the responses because they can create a dysfunctional environment for themselves and the child if they do. These responses are:

- Denial – it is hard to process the news, but when your child is sick it is hard to deny it. Once you accept it, you can manage it.
- Fear – So many unknowns and so many scary possibilities. But you must face the fear so that things don't get worse.
- Anger – Angry at the disease, the doctors, each other.
- Over-Involvement – Wrongly feeling that the child can never be left with anyone else. But you must educate others and trust them so that you do not become overwhelmed and exhausted.
- Guilt – Wrongly feeling that you are a failure or that you are to blame for it.
- Sorrow/Sadness – Very understandable, but you mustn't let your child see this in you or ever make them feel that *they* make you sad.

Laurie described some of the “daily living issues” that a family with a galactosemic child might encounter. These include:

- **Family Reorganization** – Household duties and routines might shift around
- **New Family Member** – It is just like adding any new baby, only more-so
- **Lifestyle** – Try to maintain normalcy. You risk missing out on the joy of your child's overall development if you are too focused on Gal-1-P levels, etc. Don't try to isolate the entire household from dairy products in the world. Give the child the experience of being around dairy and learning that while they cannot have these items, but that they need not fear them.
- **Siblings** – It's not the sibling's responsibility to take care of the child with galactosemia. It is vital to not create any feelings of guilt in the other siblings and therefore create a dysfunctional lifestyle. It is important to be equally aware of the needs of all children, not just the galactosemic child.
- **Loss of Privacy** – You may feel like your family lives in a fish bowl because every professional in your life is aware of personal information.

Laurie next described that a family's “Galactosemia team” should ideally include both the primary care pediatrician, and the metabolic clinic. She described that at the Denver Children's Clinic where she works, they do the blood draws, the urine-galactitol test, measure the child's weight and height, and education sessions for all ages with cooking and games.

Laurie shared that parents can also help your child by talking with them at the grocery store about “yes” and “no” foods. This can help both the parents and the children to become more comfortable with managing the diet. Laurie encouraged us that we will feel emotionally responsible when we tell our child that “this is a ‘no’ food and you can't have it and you can never have it” and when we learn to provide substitutes and alternatives so that our child doesn't feel left out.

## **Nutrition Intervention**

Laurie broke down the first five years of a child's life into different sections and commented on them individually.

### Birth to 4 Months

This is the time for parents to begin to educate themselves about how they will feed their child. Such things as learning to read labels to identify dairy ingredients and to look at lists of galactose contents in foods so as to make informed choices about inclusion of non-dairy foods in the diet.

### Four to Six months

The child needs to be developmentally ready to introduce solid foods, which is usually the case by 6 months old. There is a window of time during which you must start in order to avoid the child developing feeding issues. So par-

## Diet: Birth to Five Years (continued)

ents must not wait too long to start because of their own fears. If the child has a feeding tube, they will have an aversion to eating and it is hard to get them to eat solid foods, so you need to start solids earlier. If they have had a trauma at birth, you need to start slowly as they sometimes have a hard time.

There are two schools of thought about which foods to start first, fruits or vegetables:

1. Start vegetables before fruit so they will eat the vegetables
2. Start fruit before vegetables to them excited to eat

It's an individual decision; Laurie tends to recommend starting vegetables first and advises parents to start each new food every three days in order to see the child's reaction to each.

Parents need to plan sequencing of solid foods and should use suggested meal patterns. Eating together provides a great opportunity for education and provides a natural setting for teaching children about the differences between galactosemic and non-galactosemic diet at a young age.

During this timeframe, parents need to begin to educate siblings, extended family members, and friends about what your child can and cannot eat. When parents share this information, it might make them feel vulnerable, but they need to do it in order to start to build a support system. Parents will learn which people they can count on to 'get it' and who they can trust to help out.

### Seven to Twelve months

Laurie indicated that this period of time is ideal for parents to get some cookbooks and begin to plan meals when the child switches from baby food to table food. Isomil has a cookbook and PGC gave one to every family that attended the conference (it is available on the PGC website, too). This is a great age to start teaching "yes" and "no" foods. For the first birthday party cake, some grocery stores have dairy-free sugar icing that they will use to ice a cake. When the child begins drinking from a cup, Laurie strongly recommends that the parent replace baby formula with a calcium drink (soy milk, rice milk, Dairyfree by Vance's, etc.) so that the child gets the added calcium they need and gets use to drinking their milk substitute this way. Do not use water or juice in the cup first. Usually the child should be off of formula by two years old and definitely by three and four years old. Some people say "no" to soy milk, but Laurie has not seen any problems with it. Laurie indicated that the calcium is more absorbable in Rice Dream.

### 13-35 Months

Laurie reminded about how good parenting skills are very important in this age group. The diet of a "mobile child" will be best influenced by positive language ("You can have *this* instead of that") and minimization of 'over-involvement' by parents (start helping child to be increasingly independent. It helps to have a special bin, or drawer, or cabinet for the galactosemic child with their snacks in it. Parents must tell teachers and everyone involved in the child's care everything that is going on with your child's diet restrictions.

### Three to Five Years

Laurie described how many parents will deal with an "egocentric" child or a "negotiating" child who begs, "Can I have just one?" – the answer must always be "no." She suggested that is good to role-play and teach the child to say "I have to ask Mommy, but thank you." Laurie cautioned that a child will take food from people they trust, so it is important for parents to be mindful of who is giving food to the child. Allergykids.com is a good website resource for parents of children in this age group. Parents can put stickers on food items that might help people to stop and think before they give any food to your child. This is also the best age to start playing games that are educational and fun:

- *Fishing for Galactose*: magnetic fishing rod attaches to pictures of food. When you catch one ask is it a "yes" or "no" food. Give a positive reinforcement for correct answers.
- *Red Light / Green Light*: traffic light. Put food on light color. Is it green for "yes" food or red for "no" food. Around age 6 or 7 should get the yellow for "maybe" food.
- *Box Spins*: Put food pictures on each side of a cube. When you roll the box, say if it land son a "yes" or "no" food.
- *Drawing Food*: as they get older, draw the food and put them on green and red plates for "yes" and "no" foods.

Laurie concluded by cheerfully encouraging us that galactosemia is only one part of our child; that our children are made up of many qualities. Just as galactosemia is just a small piece of the whole person they are, it shouldn't overwhelm the way parents think about their child.

**Notes from Conference Presentations — Breakout Session**  
**Diet: Elementary School Ages**  
by Barb Bense

**Presenter:** Laurie Bernstein MS RD FADA

Laurie started off by retelling the "Welcome to Holland" story (see PGC website to read the story) and reminding us that even though we didn't plan on having a galactosemic child, our children are "very special" gifts.

Having our children going off to Elementary School can create a lot of fear for us parents because now our children are spending more time AWAY from their parents and more time with other people. Laurie told all the parents that when she was doing the training with the adolescent children, they told Laurie that their parents needed to "get a life." Laurie advised the parents not to be so overprotective and try NOT to focus our conversations with our children on "What did you EAT today?" She recommended that we treat our children) as normal as possible.

As science progresses, we have to progress as well. For example, over 15 years ago we didn't allow our kids to eat peas.

Diet is easy to manipulate, but kids learn quickly if one parents gives in easier the child will obviously go to that parent if it means he/she will be allowed to eat something. Our children aren't born with a handbook. Although, Laurie did remind the parents that because our children are comfortable saying "no" to so many foods; our kids will probably have an easier time saying "no" to drugs and alcohol as well.

Remember: Galactosemia is only a part of our child's life.

Laurie stressed the importance of educating our children about the Yes/No foods, what galactosemia is, and how to treat your child as a normal child with various activities and games. Here are a few activities and/or tips that Laurie shared with the parents:

- Make a bookmark with your thumbprint and decorate with their favorite things! This activity shows everyone how different EVERYONE is no matter if he/she has galactosemia or not.
- Trace the body of someone on a large piece of paper and then show them WHERE their liver is. (Laurie also shared that she has actually brought in a liver from an animal so the kids can see one and see what it feels like. The liver is in a baggie, of course.)
- When sending a lunch with your child, explain to your child what exacting you are sending so that he/she knows what you're sending and that it is a YES food.
- Use the red/yellow/green light for food with children aging from 6 – 9 years old. (Red means NO foods, Yellow means MAYBE foods, and Green means YES foods.) When children are between 9 and 11, they should be able to learn more of the YES, NO and MAYBE ingredients.
- You can make a book of pictures of YES, NO and/or MAYBE foods with your child. One parent told the group that her child brought the book to school to show her teachers.
- Talk to your children about the importance of calcium.
- Talk about "Ways We are Alike" and "Ways We are Different" with children.
- Always remember to reward your children. It doesn't always have to be with treats/food.
- There's a website ([www.AllergyKids.com](http://www.AllergyKids.com)) that is selling a symbol (!) so that it creates an awareness/reminder that your child has serious food allergies. For more information, go to their website.
- Make learning fun! (This is VERY IMPORTANT!)

**Notes from Conference Presentations — Breakout Session**  
**Diet: Adolescent Ages**  
by Kelly Salvetti

**Presenter:** Laurie Bernstein MS RD FADA

Laurie was asked to conduct this breakout session to discuss the issues and problems unique to the Galactosemic adolescent's diet. Prior to conducting this breakout session, Laurie attended a session with "Generation G", young adults in their late teens and twenties with galactosemia. She was so "blown away" (her own words) with the feedback she received from this group of kids that she abandoned her formal presentation to this group in order to share with us the things she learned from the Generation G kids.

Adolescence is defined as early (11-14 y/o), middle (15-17 y/o), and late (18-21 y/o). Our Galactosemic adolescents are in many ways no different from other adolescents their age. They are all struggling to gain independence from their parents while still desiring to be "taken care" of sometimes. Adolescents desire and need to have privacy from their parents and other significant adults in their lives. Adolescents are facing the future and have anxiety about planning short and long-term goals.

Adolescents who have Galactosemia have to contend with all of these things, but have additional obstacles to overcome. All children with Galactosemia are accustomed to their special dietary restrictions. As children grow into adolescence and spend more time away from "mom" they are placed into situations when it is uncomfortable to be different. For example; your child goes with a group of other kids to Pizza Hut after a football game. Your child begins to have dialogue with the food server about the milk content in the pizza dough, when the cutest girl or boy that he or she has ever seen walks in. "Generation G" told Laurie that they would abandon the "ingredient dialogue" and just go along with the flow; to the point of eating an "unsafe" food in order to save themselves embarrassment. Of course, this is "risk-taking" behavior.

Generation G spoke a lot about dying. They were very concerned with accidentally eating something bad for them with death being the consequence of that action in their minds. There was a lengthy, animated conversation among the participants of the session about why the kids were so concerned about dying.

For many of us parents, when our children were diagnosed with Galactosemia, we felt like our children would die if they ingested milk. After all, some babies have died and others were so sick it was unsure if they would survive their illness. As parents, we wanted to protect our children and some of us told our children they would die if they drank milk. In retrospect and as our infants grew to be older children, we gained more understanding of Galactosemia and understood that it would be the long-term, repetitive ingestion of milk that would cause the damage. Evidently, the children haven't come to the same conclusion, because they all expressed fear of death at the accidental ingestion of an "unsafe" food. Even though, given the right circumstances they might eat it in order to avoid embarrassment.

It is very important that we talk with our children and instruct them correctly. It's important to speak with them about risky behavior, but make sure they know that death is not the immediate consequence of eating "unsafe" foods. It's important to speak with them honestly and very directly.

Generation G asked a lot of questions about what happens when they have children. It is important to discuss the genetics of Galactosemia with them. They are unaware of the percentage of chance that they will have a child with Galactosemia.

Generation G desires to gain autonomy from their parents, but some of these kids have unique challenges to overcome. The kids don't like to be labeled with a "disability". It is better to refer to any difficulties as "challenges" instead. The kids shared that school is very hard for them. Other kids were described as "mean" and high school was described as "terrible". The kids felt misunderstood by their parents. Sometimes the kids said "we try so hard"; "I can't help it", "people aren't nice to me". Some kids expressed that they tried as hard as they could, but just couldn't master some things and their parents didn't understand that they just couldn't do it.

Some of our children have deficiencies with reading and writing. This makes it very difficult for them to gain independence from their parents. It's important to try to give them the tools they need to gain autonomy; help to make it easier to do things on their own. Some ideas were to provide them with recipes, visual instructions in place of

written instructions, making needed items accessible, locating outside agencies that may be helpful, etc.

Communication should be concise, direct, age-appropriate, and clear. It's important to teach them the reality of Galactosemia and the known complications. As these children grow older, it becomes important to help them bridge the gap between dependence on you and self-sufficiency. They may need help locating medical insurance and determining what insurance will and will not pay for. Some young adults may not qualify for medical insurance and need help finding an alternate means of paying for medical care.

Some things to consider teaching your adolescent: Diet for Life – Biochemistry; adverse effects of being off of diet; practical applications to diet management; current thoughts about family planning and recurrence risk (carrier testing).

The last subject pertains to girls. Don't wait until a girl is 18-19 years old to have a conversation with her about premature ovarian failure. The girls tend to be angrier and have a harder time accepting if they are not introduced to the possibility sooner. The decision to talk to your daughter about this is of course dependent upon age and maturity.

### Notes from Conference Presentations — Breakout Session **Topics in Galactosemia** by Kelly Salvetti

Presenter: Dr. Louis Elsas, M.D.

Galactosemia is one of the most common carbohydrate metabolism disorders. Most cases of classic Galactosemia are caused by the absence of the enzyme galactose-1-phosphate uridylyltransferase (GALT). The enzyme deficiency is an autosomal (non-sex chromosome) recessive genetic condition.

Most states perform newborn screening tests and Galactosemia is identified by a positive or abnormal result. Untreated infants typically present with lethargy, hypotonia, jaundice, liver dysfunction, bleeding, sepsis, cataracts, and failure to thrive to death. Unfortunately, even those infants with an early diagnosis become ill and require hospitalization with intensive care for several days. Early diagnosis is within 5-7 days, but it can take as long as 2 weeks to obtain results. Meanwhile, infants suffer from hepatocellular failure, e. coli sepsis, and death. Long term complications include cataracts, dyspraxia, ovarian failure, neurological dysfunction, and growth and development delays.

Dr. Louis J. Elsas and his colleagues from the Dr. John T. MacDonald Foundation are working to develop a new method of newborn screening. This method would use  $^{13}\text{C}$ -labeled substrates and quantify  $^{13}\text{CO}_2$  in expired air. This test would be performed on a newborn while still in the nursery. It is safe and provides rapid, accurate functional assessment of whole body galactose oxidation.

There are two goals for this new screening method: 1) validate and optimize the  $^{13}\text{C}$ -Galactose Breath test in newborns; apply the breath test to healthy newborns and determine age related changes 2) develop an inexpensive, point of care spectrometer to measure  $^{13}\text{Co}_2/^{12}\text{Co}_2$  ratios.

Genotype is defined as the "internally coded, inheritable information" carried by all living organisms. This stored information is used as a blueprint for building and maintaining a living creature. These instructions are within almost all cells and are written in a coded (genetic code) language. They are copied at the time of cell division or reproduction and are passed from one generation to the next. These instructions are involved with all aspects of the life of a cell or an organism.

An allele is one member of a pair or series of genes that occupies a specific position on a specific chromosome.

Most children with classic Galactosemia have a Q188R mutation on one or two allele(s). There are other allele(s) associated with Galactosemia such as L195P, V151A, S135L, etc. Research has shown that those people who are homozygous (have two of the same alleles) for the Q188R mutation have much less expired  $^{13}\text{Co}_2$  than those who have different variations of the mutation.

There is no difference in calculated cumulative percent dose of expired  $^{13}\text{Co}_2$  in normal newborns in their first 2 days of life. We're not sure what, but something happens after day 2 to "turn on" galactose oxidation.

## Topics in Galactosemia (continued)

The breath test is also a good predictor of dyspraxic speech and ovarian failure in older children with Galactosemia.

In patients with classic Galactosemia there is a disruption of normal protein synthesis caused by an accumulation of Galactose-1-phosphate.

- a) Galactose is metabolized to Galactitol and Galactonate (both of which are excreted in urine).
- b) Galactose is metabolized by Galactokinase (GALK) to Galactose-1-Phosphate (this is what accumulates in the red blood cells of classic galactosemics) and UDP-glucose (essential for all protein synthesis).
- c) Gal-1-P is in turn metabolized by the enzyme Galactose 1 phosphate Uridyltransferase (GALT) (missing enzyme in classic Galactosemia) to Glucose-1-Phosphate and UDP-galactose.
- d) Glucose-1-Phosphate translates into energy metabolism.
- e) UDP-glucose and UDP-galactose is metabolized by UDP-galactose-4-epimerase (GALE) to glycolipids and glycogen (biosynthesis).

Elevated accumulations of Gal-1-P reduce UDP glucose and UDP galactose processing and alter post translational processing of proteins. Messenger RNA encodes and carries information from DNA during transcription (the writing of proteins to a ribosome) to sites of protein synthesis to undergo translation in order to yield a gene product. Patients with classic Galactosemia who have elevated Gal-1-P levels undergo "abnormal" translation (post-translational processing) of proteins to the cell. It is thought that this is what causes some of the long-term complications we see with classic Galactosemia.

An endoplasmic reticulum stress response related to GALT deficient cells also causes cells to die in the brain, liver, and ovaries.

It is the hope of Dr. Elsas that the breath test can be perfected and transformed into an affordable alternative to the current newborn screening test. If we can detect and diagnosis Galactosemia sooner we may be able to change the adverse outcomes associated with classic Galactosemia.

## Dr. Stanton Segal's Research at CHOP

The Metabolic Research Laboratory at CHOP is focused entirely on understanding galactosemia. It is the premier research unit in the world devoted to projects that can benefit those with this disorder. Dr. Segal has over 50 years of experience studying galactosemia and has published over 100 clinical and basic research papers related to galactosemia. Our current projects include studies on:

1. How classic galactosemics metabolize galactose
2. The amount of galactose that older people with galactosemia should have in their diet
3. What drugs may depress galactose metabolism
4. A comparison of Isomil vs Neocate in the treatment of newborn galactosemics
5. Brain metabolism studied by PET and functional MRI techniques
6. Why mice with absent galactose-1-phosphate uridyltransferase (GALT) show no symptoms of galactose toxicity when fed galactose

All of these studies are in danger of being interrupted because of decreased funding for research by the NIH (National Institutes of Health) and the possible closing down of the laboratory.

Funding is needed for all of these projects and we are currently applying for multiple research grants. However, we are asking for your help in fundraising for the last project that needs immediate support. The study involving the GALT-deficient mice that have no symptoms may lead to a new approach toward galactosemia, using medication. We have found out that these mice make high amounts of gal-1-P but no galactitol in target organs as humans do. We have made mice by genetic manipulation that can make galactitol in the ovary and liver. When these are bred with our GALT-deficient mice, we expect to see ovarian and liver toxicity, as these accumulate both gal-1-P and galactitol. If this is the case, it may be possible to prevent this with available drugs called aldose reductase inhibitors. These drugs have already been developed and are currently used in the treatment of other disorders. This could lead to medications being prescribed for people with galactosemia that would stop galactitol formation. With less galactitol, there is a possibility of preventing long-term complications. Stopping this project at this point in our progress would be a great blow to not only the future knowledge of galactosemia but to possible new medication usage. With the data from our mouse study, the FDA could be approached for approval to perform a clinical trial on these drugs in individuals with galactosemia

Dr. Stanton Segal at The Children's Hospital of Philadelphia (CHOP) is conducting a study to compare Isomil which contains small amounts of galactose with Neocate which has no galactose in the treatment of newborn galactosemic infants under the auspices of NIH. The study involves placing newly diagnosed babies on either of the infant formulas with evaluations at monthly intervals for 6 months in the CHOP Clinical Research Center for growth and development and the measurement of galactose and galactose metabolites RBC gal-1-P, galactitol and galactonate, plasma galactose and galactitol and urinary galactitol and galactonate. If you are interested in having your baby participate in our study and you live within a reasonable distance from Philadelphia, please contact Dr. Segal at 215-590-3372 as soon as your baby is identified as having galactosemia.

## Introducing New Regional Galactosemia Support Groups

### **Galactosemia Support Northwest**

3800 NE Sandy, Suite 125  
Portland, OR 97232  
[www.galactonw.org](http://www.galactonw.org)

#### **About us**

Ron and Beate are the parents of Alena, 2-1/2 and Mia Rose 4 months. Both children have Classic Galactosemia. When Alena was first diagnosed in 2003 Ron and Beate were desperately looking for a local support group, as they were frightened and did not know what to expect. Desperately they began a search for parents or patients living near them to provide information. They found little support available on the West Coast. As a result, Ron and Beate Krull, along with two friends started Galactosemia Support Northwest.

We are very excited about the launch of Galactosemia Support Northwest. For more information, please feel free to e-mail Beate [info@galactonw.org](mailto:info@galactonw.org) , or call 503.236.6838

#### **Goals of Galactosemia Support Northwest**

It is our goal to facilitate communication between those affected by Galactosemia and to provide support for individuals and families living in the states of Alaska, Hawaii, Idaho, Montana, Oregon, Wyoming, and Washington. The primary goals of the organization are to provide funds for research and raise awareness of the disorder. In addition, we will provide recipes, food ideas, a restaurant guide for the greater Portland area, travel tips including tips for travel abroad, and will be a liaison to Galactosemia support groups in other countries.

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### **Galactosemic Families of the Southern States**

Calling all Texas, Louisiana, Oklahoma and Arkansas families.

We are starting a new group called. GFSS planned to hold their first meeting on Saturday September 30, 2006 at 1:00. It was held at Children's Medical Center of Dallas. If you have any questions or are interested in helping out, email Amanda Rawls at [Amandarawls@sbcglobal.net](mailto:Amandarawls@sbcglobal.net)

## PGC's Operating Income and Expenses (1/1/06—9/18/06)

INCOME		EXPENSES	
<b>2006 Conference Income</b>		<b>2006 Conference Expenses</b>	
2006 Conference Back Packs Sold	\$40.00	2006 Conference Backpacks	\$375.00
2006 Conference Donation Cup	\$100.00	2006 Conference Badges	\$269.96
2006 Conference Petty Cash	\$600.00	2006 Conference Banquet AV	\$7,292.26
2006 Conference Registration	\$20,437.00	2006 Conference Baseball Game	\$1,155.00
2006 Conference Shirts - Philly	\$105.00	2006 Conference Food	\$17,753.30
2006 Conference Sponsor	\$1,250.00	2006 Conference GG Program	\$353.06
2006 Conference Tote Bag Donation	\$600.00	2006 Conference Honorariums	\$711.30
2006 Conference Tote Bags Sold	\$40.00	2006 Conference Lodging	\$8,379.00
<b>TOTAL 2006 Conference Income</b>	<b>\$23,172.00</b>	2006 Conference Office Supplies	\$457.21
<b>Bank Fee Refund</b>	<b>\$133.00</b>	2006 Conference Petty Cash	\$600.00
<b>Donations</b>		2006 Conference Postage	\$334.22
Kyleigh Lydon's Cure	\$1,105.00	2006 Conference Printing	\$2,272.29
Sarah Southard's Cure	\$3,697.00	2006 Conference Refund-Overpayment	\$130.00
Other Donation	\$17,923.99	2006 Conference Scholarship	\$443.33
<b>TOTAL Donations</b>	<b>\$22,725.99</b>	2006 Conference Shirt (Philly)	\$1,337.00
<b>Fundraising Income</b>		2006 Conference Tote Bags	\$962.50
Fundraising - "Not Milk" Tee Shirts	\$975.00	2006 Conference Travel	\$2,393.67
Fundraising - Aqua Tee Shirts	\$435.00	2006 Conference Youth Program	
Fundraising - Cafepress	\$358.97	2006 Conference Teachers-Childcare Wkrs	\$1,396.20
Fundraising - Can Huggers	\$72.00	2006 Conference Youth Program	\$1,594.55
Fundraising - Lunch Bags	\$100.00	<b>TOTAL 2006 Conference Youth Program</b>	<b>\$2,990.75</b>
Fundraising - Magazine Sales	\$700.00	<b>TOTAL 2006 Conference Expenses</b>	<b>\$48,209.85</b>
Fundraising - Recycle	\$13.00	Accounting	\$974.00
Fundraising - Silicone Bracelet Sale	\$676.00	Bank Charge	\$203.00
<b>TOTAL Fundraising Income</b>	<b>\$3,329.97</b>	Finance Charge	\$11.05
Interest Income	\$258.43	Fundraising Expense	
<b>Memory Donation</b>		Fundraising - "Not Milk" Shirt	\$360.00
In Memory Myrna Kaufmann	\$10.00	Fundraising - Buttons	\$141.10
In Memory of Leon Bobis	\$875.00	Fundraising - CafePress	\$191.50
<b>TOTAL Memory Donation</b>	<b>\$885.00</b>	Fundraising - PGC Aqua Shirt	\$720.50
Opening Balance 2006	\$63,959.91	Fundraising - PGC Can Hugger	\$150.00
<b>TOTAL INCOME</b>	<b>\$114,464.30</b>	Fundraising - PGC Lunch Bags	\$400.00
		<b>TOTAL Fundraising Expense</b>	<b>\$1,963.10</b>
		Licenses and Permits	\$217.86
		Office Supplies	\$539.65
		Postage and Delivery	\$209.37
		Printing and Reproduction	\$905.22
		Transfer to Research Account	\$50.00
		Travel, Business	\$652.13
		Telephone-Toll Free Max	\$186.31
		<b>TOTAL EXPENSES</b>	<b>\$54,121.54</b>
		<b>TOTAL BALANCE FORWARD</b>	<b>\$60,342.76</b>

## PGC's Research Fund Income and Expenses (1/1/06—9/18/06)

<b>INCOME</b>	
Bank Fee Refund	\$1.00
Donation	
Center of Excellence Donations	\$1,000.00
Kyleigh Lydon's Cure	\$90,100.00
Other Donation	\$1,900.00
<b>TOTAL Donation</b>	<b>\$93,000.00</b>
Interest Inc-Interest Income	\$73.13
Memory Donation	
In Memory Harold Wagener	\$200.00
<b>TOTAL Memory Donation</b>	<b>\$200.00</b>
<b>TOTAL INCOME</b>	<b>\$93,274.13</b>
<b>EXPENSES</b>	
Bank Charge	\$1.00
Bank Supply Fee	\$24.56
Center of Excellence Expense	\$70,000.00
<b>TOTAL EXPENSES</b>	<b>\$70,025.56</b>
<b>Previous Balance Forward</b>	<b>\$2,241.05</b>
<b>OVERALL TOTAL</b>	<b>\$25,489.62</b>

### PGC Funds Research and a Whole Lot More.

With the long-pursued goal of a centralized center for galactosemia research becoming a reality, PGC will finally be able to focus fundraising efforts on providing support to sustained research. The PGC board stands committed to providing as much support to this center of excellence as is possible. Direct financial support to the center will likely be PGC's single largest line item expenditure every year for the foreseeable future. And fortunately, our donations will give us a voice in helping to direct the course of the center's activities. But the Children's Hospital Boston Galactosemia center will not be our only expenditure.

It is vital that PGC's general operations account remain well-funded in order for the organization to remain viable. As you can see from the expense report, less than half of the conference cost was paid for by the attendees' registration fees. More than half of the conference expenses were paid for with prior-raised funds from the general operations account. Fortunately, that account's balance forward is sufficient to keep PGC growing healthily. If all the group's fundraising is directed towards research, there will be dwindling funds left over for conferences, newsletters, and other programs. So please consider splitting your future donations to both research and operations. Perhaps you would feel comfortable with a 90% research / 10% operations split, or 75/25, or even a 50/50. You can be certain that your donations will be directed to the accounts exactly as you specify.

Remember, too, that 100% of PGC's activities are performed by unpaid volunteers. 100% of donated funds in the research fund go to fund research. 100% of funds in the general account go to pay for goods and services necessary to conduct PGC's efforts.

The same holds true for support of regional galactosemia groups. It is wonderful that these groups can form to have a more 'hands-on' support among families at a local level. Hopefully, funds that they raise in excess of their local needs can be directed to PGC as a form of grass roots fundraising support. For example, the GANES group paid for the Dessert Social at the recent conference. Direct support to PGC like this really helps PGC's budget go further.

PGC has been blessed by the generosity that so many of you have shared with PGC in the past. All of PGC's volunteer board members, committee chairpersons, and committee members remain dedicated to being good stewards of the organization's resources in pursuit of our shared mission and values. Please continue to support our community mission of support, education, and research with your donations.

## Notes from Conference Presentations — Breakout Session "Supporting Your Child's Sensory Preferences" By Beth McVicar

**Presenter:** Linda Beck

Linda Beck is an occupational therapist at Orchard Friend's School. Orchard Friend's School is a school dedicated to the education of children with language-based learning differences. Linda described the basics in sensory integration. It is a feeling that "something just does not feel right" or "a sense of not feeling comfortable". The different senses, hearing, touch, sight, taste and smell all have receptors that communicate with the brain and can formulate a response. There are also two hidden senses, muscle sense and balance sense. It is difficult to do fine motor tasks, for example if you are not getting proper input to your muscles. It is difficult to sit quietly and concentrate if your balance sense is off. Children can be overloaded with too much input. The nervous system is not able to be a good traffic cop. When senses are integrated the nervous system can handle all the input and the brain can issue the appropriate response.

Linda discussed how to change your alert level. Your body is like a car engine, sometimes you may feel like your engine is running in high speed, in low speed or just right. When your engine is in high gear, you may find it difficult to pay attention, to sit quietly in your seat, or get your work completed. When your engine is in low gear, you also may find it hard to concentrate, you may "daydream" easily, or feel like a "couch potato." When you are in the "just right" place, it is usually easier to pay attention, to get your work done, and to have fun.

If you want to change your engine gear from high or low to get into the "just right" feeling you may want to try the following:

- **Put something in your mouth:** eat hard candy, eat crunchy foods, eat chewy food, eat sour food, eat sweet food, drink from a straw, take slow deep breaths.
- **Move:** try moving before you need to concentrate – ex homework, walk quickly, run up and down steps, do an errand for a teacher, use a therapy ball
- **Touch:** try holding and "fidgeting" with a Koosh Ball, paper clips, rubber bands, straw, rub gently on your skin or clothing, wash your face with a cold or hot wash cloth, hold or learn up against a stuffed animal or large pillows
- **Look:** put bright lights on in room if you are in low gear, dim the lights if you are in high gear, clear off the table you are working on if it distracts you, watch fish in an aquarium, read a book or look at magazine
- **Listen:** listen to classical type music (even, slow beat), listen to hard rock type music (loud bass, uneven beat), use a personal cassette player if the music bothers someone else, avoid loud, noisy places if you are in high gear or if it bothers you when you are trying to concentrate

Linda also discussed Sensory Strategies for Parents – Behaviors observed at home:

- **Observation:** Clumsy/accident prone, bumps into things or people, breaks things  
**Possible Sensory Cause** - Difficulty judging body position, not getting enough sensory information to muscles and joints.  
**Sensory Strategy** – Provide more proprioceptive input: weighted items (vest, ankle or wrist weights).
- **Observation:** Dislikes being hugged or kissed.  
**Possible Sensory Cause** - Unexpected touch may be uncomfortable.  
**Sensory Strategy** – When appropriate, allow the child to determine when he will be hugged or kissed. Prepare family members or friends of the child's preference. Let the child know before a hug or kiss takes place.
- **Observation:** Prefers engaging in sedentary activities.  
**Possible Sensory Cause** – Limiting actions may avoid unpleasant sensations or unpredictable movement. May have difficulty motor planning. Anxiety about performing.  
**Sensory Strategy** – Embed physical activity into routine tasks- making bed, taking out trash. Incorporate physical activity into sedentary activity-get game from high shelf. Walk up stairs to get to game.

## **"Supporting Your Child's Sensory Preferences"**

*(continued)*

- **Observation:** Messy Eater:  
**Possible Sensory Cause** - Not receiving enough tactile input to be aware of food on mouth/face. May have difficulty managing utensils.  
**Sensory Strategy** – Massage lightly around the child's mouth with different textures: soft washcloth, soft toothbrush. Try activities/games that involve the mouth-bumbles, straw, kazoo.
  
- **Observation:** Dislikes certain clothing-only wears cotton, old sweatshirt, etc  
**Possible Sensory Cause** – Tactile Sensitivity  
**Sensory Strategy** – Remove tags if bothersome Massage the child with a vigorous towel rub to increase tolerance to certain fabric textures Consult an OT re: Wilbarger Protocol
  
- **Observation:** Trouble dressing-buttons, snaps, etc.  
**Possible Sensory Cause:** Difficulty with fine motor skills. Weakness in hands.  
**Sensory Strategy** - Start with large fasteners that are easier to manipulate. Use Velcro. Cue child to look at the fastener. Provide opportunities to strengthen muscles in hands (play-doh, light brite, draw on chalk board, pick up things with tongs, etc.)

### **Behaviors Observed at School**

- **Observation:** Chews on clothing, pencils, etc.  
**Possible Sensory Cause** - Seeking proprioceptive input to jaw, oral tactile input  
**Sensory Strategy** - This may be helping the child to focus and calm—try other items that are more appropriate, Sugar-free gum, straws, coffee stirrers, water bottle with straw. Provide crunch and chewy snack / lunch.
  
- **Observation:** Has difficulty switching classes in the hallway.  
**Possible Sensory Cause** - Sensitive to unexpected touch. Loud noises / volume Visual input / movement. **Sensory Strategy** – Leave class a few minutes early. Hold door open with back to get more proprioceptive input. Ask for locker to be at the end of row, less contact with peers.
  
- **Observation:** Notices every little sound or visual change in the environment.  
**Possible Sensory Cause** – May have trouble discriminating important sounds and sights from unimportant.  
**Sensory Strategy** – Be conscious of the environment and limit visual and auditory distractions. Establish a quiet area with preferred sensory support. Consider a visual barrier. Provide headphones or earplugs to use during testing or seatwork.
  
- **Observation:** Has messy handwriting: unable to stay within the lines.  
**Possible Sensory Cause** – May not be receiving appropriate sensations to form motor response. May not have an adequate pencil grip.  
**Sensory Strategy** – Gross motor warm up to writing or coloring: wheelbarrow walk, push against wall or person with arms outstretched, clapping hand games. Darken the bottom line, “this is where the letters sit.” Air write the letter or word.
  
- **Observation:** Difficulty keeping hands and feet to self.  
**Possible Sensory Cause** – Seeking tactile input. Doesn't have a sense of personal space.  
**Sensory Strategy** – Visual or physical boundaries – tape, carpet square, cushion, hula hoop. Sit in adults lap (if appropriate). Use a finger fidget. Lie on stomach while propped on elbows.

*Looking for a Perfect Holiday Gift ? Introducing the ...*  
**Galactosemia Awareness Bracelet**

This bracelet was designed by PGC President, Michelle Fowler, in an effort to further awareness of galactosemia and the effects it has on those children with this rare disorder.

Children with galactosemia are missing the *GALT* enzyme which is normally made by the ninth chromosome of DNA. In the bracelet, this enzyme-producing location is represented by the nine Swarovski colored crystals. The bracelet's other colored crystals represent qualities that we wish for our children and all who live with galactosemia to possess. (see PGC's website for picture of a bracelet)

Violet signifies **self-esteem** that we strive to instill in our children, but which they sometimes lack due to living such a challenging life-style.

Blue signifies **sincerity, and health** that those with a normal way of life must never take for granted.

Jonquil signifies **hope** that, through research, a cure and better way of life for those with this disorder will be found.

The heart charm symbolizes the power of love - a love that reminds us that all people deserve compassion and understanding. To emphasize the uniqueness of each individual with galactosemia, the heart charm on each bracelet is different. Please accept the heart you receive just as you would embrace and love the unique gifts each child with galactosemia brings to the world.

NAME \_\_\_\_\_

ADDRESS \_\_\_\_\_

CITY \_\_\_\_\_ STATE \_\_\_\_\_ ZIP \_\_\_\_\_

Bracelets

# of bracelets \_\_\_\_\_ size 6 ¾" @ \$ 30.00 ea. \_\_\_\_\_

# of bracelets \_\_\_\_\_ size 7 ¼" @ \$ 30.00 ea. \_\_\_\_\_

# of bracelets \_\_\_\_\_ size 7 ¾" @ \$ 30.00 ea. \_\_\_\_\_

Shipping fee \$ .75 per item \_\_\_\_\_

TOTAL \_\_\_\_\_

Please make checks payable to:

PGC  
P.O. Box 2401  
Mandeville, LA 70470-2401



*Bringing Families Together with Experts and Each Other*

**PARENTS OF GALACTOSEMIC CHILDREN, INC.**

Parents of Galactosemic Children, Inc.  
P.O. Box 2401  
Mandeville, LA 70470-2401

Toll Free Phone: 1-866-900-7421  
Email: [president@galactosemia.org](mailto:president@galactosemia.org)

## **New on PGC's website**

Those of you who have been to PGC's website at [www.galactosemia.org](http://www.galactosemia.org) recently have noticed big changes. Our website was redesigned by David Reid, father of a son with classic galactosemia. His volunteer effort has given our website a polished and up-to-date look. But even more importantly, he has set in place the means for the website to grow and expand in the future.

It is not just the 'look and feel' of the website that has changed. We have added new content of interest to the community. Many of the conference handouts are there for your review. There are new items with the PGC logo to purchase, new opportunities to help us raise funds by recycling printer cartridges, a database of physicians who see galactosemia patients, the recipe book from the conference, and the 'our stories' booklet from the conference.

In the near future, we will expand the website even more to bring back the roster of families, accept donations via credit card, allow for conference registration by credit card, and provide the slides from speakers at the conference.

PGC would also like to thank volunteer website host, Scott Scull, father of a daughter with classic galactosemia. Without his generous donation of time, and resources, PGC would not have our own little corner of cyberspace to call home.