

The Galactosemia Gazette

Volume 3, Issue 1

Spring 2007

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- POF (parent)

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PGC's Research Program Preparing to Launch

Therese Cozzo
Chairperson, PGC Research Committee

Two years ago, PGC established a research fund and began accepting donations specifically for the purpose of funding research into areas of interest to families living with galactosemia. With this fund, PGC has already made donations to general galactosemia-related research projects or centers.

In the interest of directly funding *specific* research projects of interest to the PGC community, the Research Committee is currently working to establish a Research Grants Program by which PGC will accept research proposals and fund them on an individual basis, based on both their scientific merit and the interest of the PGC community. In this way, the families living with galactosemia may have a larger voice in galactosemia research.

Once the Research Grants Program is established and approved by PGC's board of directors, it will be published on the PGC web site. The Research Committee will also publish a brief description of each winning proposal and will share any progress reports or final reports from funded research projects in order to keep you fully informed.

The Research Committee welcomes and encourages your input at any time. Please submit suggestions or comments regarding research areas of interest to research@galactosemia.org.

We also ask that you please consider making a tax-deductible donation to PGC's Research Fund (in addition to your donations to the General Fund) in order to help keep this research program well-funded and focused on items that matter most to families living with galactosemia. We appreciate all those who have already donated to the Research Fund. We know how precious this fund is to all who have given to it, and we intend to do our part to ensure that these donations are spent wisely.

Finally, I wish to thank Research Committee member *Christy Johnson* for her time and talents in serving this committee. I also wish to thank former members *Angela Forsyth* and *Mike Denny* for their time and contributions, and I wish to welcome our newest members: *Dan Lambert*, *Jo Beth Southard*, and *Christine Raum*.

Please Note PGC's New Mailing Address

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

If your company sends 'matching' donations, please let them know of this change

TX Fundraiser for Research is a Huge Success

Sarah's Cure, a benefit concert / silent auction, was held on October 14, 2006 on the square in historic downtown Decatur, Texas. Over \$25,000 was raised for the PGC research fund.

Our vision for Sarah's Cure was a result of our involvement in the educational program, Landmark Education. During this program, we were challenged to organize a community event that touched, moved, and inspired us. We, of course, have a huge passion for family and helping Sarah, our 7 year old daughter with classic galactosemia, lead as normal a life as possible. With that in mind, we decided to organize a benefit concert in our community. After talking to Sean and Kristine Lydon and discussing the huge success they had with their auction, we added a silent auction and live auction to the activities. With tremendous support from our family and friends we organized the 1st Annual Sarah's Cure.

Despite the rain, the benefit was a huge success. Approximately 325 people were in attendance which included 7 families that have children with galactosemia. All the families were able to get onto the stage and thank everyone for their support. I of course was speechless and in tears with appreciation to all the people that came out and supported us.

Mark and I have committed to making Sarah's Cure an annual event. This year we plan to have the fundraiser in October in downtown Fort Worth and have a goal to raise \$75,000. What we realized is that it is not that hard to make a difference in this world and there are a lot of people out there that are more than willing to help you do it. Together we can find a cure or treatment for galactosemia; I believe that from the bottom of my heart.

- Jo Beth and Mark Southard

Galactosemic Families of MN Update

The Galactosemic Families of MN (GF-MN) had their fall meeting at the Grand Lodge Hotel in Bloomington, MN on September 30, 2006. Thirteen families attended the meeting and several families stayed and enjoyed the water-park after the meeting.

We decided to send out "Welcome Bags" to the new galactosemic families. The bags included the following items: a black tote bag with the GF-MN logo, a bib with the GF-MN logo, PGC Adult bracelet, PGC lunchbox, GF-MN cookbook, "Understanding Galactosemic Guide Book" (from University of WI), "Parents Guide to Galactosemia: Parts 1 & 2" (from CA Department of Health Services), a cup-a-cake holder, a Special Diet Exemption Card (from MN Department of Health), a GF-MN brochure and a PGC brochure. In October, we mailed out eight bags to families who had children under the age of two to get their feedback. We have sent out an additional seven bags in February to new families in MN & WI.

One GF-MN family has agreed to collect and send in empty toner cartridges for GF-MN.

We will have our spring meeting on April 21, 2007, at 9:30 a.m. at the Grand Lodge Hotel in Bloomington. Our speakers will be talking about the importance of calcium and osteoporosis.

Here's a breakdown of the families on our mailing list:

- 42 families in MN
- 12 families in WI
- 4 families in IA
- 1 family in each of the following states: ND, SD, and NE
- 9 medical professionals in MN, IA and WI

NOTE: Some of the WI families may decide to attend meetings in the Chicago area once they start having their meetings.

- Barb Bense, GFMN

PGC Raised Up On Eagles Wings !!

The Eagles of Eastside High School soared for PGC last fall. The students, from a suburb of Greenville, South Carolina, made a donation of \$78,000 to PGC following a whirlwind of fundraising activity.

Each year, the students of Eastside High compete against students from their rival high school to see who can raise the most money for charity in a week-long series of events. The selection committee looks for an opportunity to help out a small charity where their work can really make a difference. Nick Elliot and Sabrina Honeycutt, Greenville area residents and parents of a six year old son, Josh, with classic galactosemia, presented on behalf of PGC. Nick and Sabrina pointed out the facts about galactosemia such as its rarity and potentially serious complications. Having had their heart strings tugged a bit by the difficulties that some galactosemics face, the Eastside students selected PGC from among a dozen or so other candidate charities to be the recipient of their fundraising largesse. Sources report that Josh's irresistible blue eyes sealed the deal.

The Eagles then got busy in their tireless efforts to raise money. In addition to traditional fundraising activities such as car washes, battle of the bands, and yard sales, the students and teachers of Eastside got creative and had some fun too. Some teachers sold 'minutes of no teaching' where students could silence the teacher with a contribution. Other teachers volunteered to shave a mustache (or even their head!) if their students could meet a predetermined fundraising level. Students also bid in an auction for the chance to push a pie in the face of teachers or administrators. The surrounding community got involved too. There was a 5K trail run through Paris Mountain State Park, an alumni dinner catered by the Atlanta Bread Company, and a 'Miss Spirit Week' Pageant. The results of the fundraising competition were announced under the Friday night lights at halftime of the big game, complete with the presentation of a 'larger than life-sized' check.

This donation is surely larger-than-life for PGC. We are grateful to Nick and Sabrina for getting PGC selected and especially to the Eastside Eagles and everyone in the Greenville-area for their hard work and support to PGC. This contribution will allow PGC to fund proposals under the research program as well as continue ongoing support activities.

New Regional Groups Forming

ROCKIES

Several families in the Colorado/Rocky Mountain Region are starting a local galactosemic families support group. Our first meeting will be held on May 21, 2007. We'll be naming the group at the meeting. Our meeting location has yet to be determined. We are meeting after our group clinic appointment at the Children's Hospital in Denver. If you are interested in joining or would like more information about the meeting when it becomes available, please contact Stacey Aurzada at staceylaurzada@yahoo.com or at 970-330-3419.

Thank you,
Stacey Aurzada - *Mom to Abby (age 4) and Ella (age 2 and CG)*

MIDWEST

Several families in the Midwest Region are starting a local galactosemic families support group as well. Their first meeting was to be held on May 19, 2007 in Roselle, IL from 2:30 to 4:30 at the public library with events planned for both adults and children. Topics of discussion include diet management, update from the PGC conference, and impact on the family. For more information, contact :

Melissa Edwards, RD
Children's Memorial Hospital
2300 Children's Plaza, Box 59
Chicago, IL 60614
Phone: 773-880-4025
Email: medwards@childrensmemorial.org

New Regional Group Formed in the South

Jo Beth Southard
President of GFSS

Galactosemic Families of the Southern States (GFSS) was formed in the fall of 2006. This regional group is a non-profit, volunteer organization that is serving families in Texas, Louisiana, Oklahoma and Arkansas. Our mission is "To provide support and community for individuals and families affected by galactosemia and to support education and research for the treatment or cure." (GFSS Mission Statement)

One immediate goal is to have an annual social event. This will allow families to develop and nurture lifelong friendships with other families who have children with galactosemia. It will also help the children and parents know they are not alone.

Our first social event will be held on June 9, 2007 at 10:00am at NRH2O, a water park in North Richland Hills, TX. We are collecting names and money early to get a group discount rate and GFSS will reserve 2 picnic areas where everyone can congregate and socialize. We will also be having a barbeque cookout, "Texas Style", at Jo Beth and Mark Southard's house in Decatur, TX on Friday, June 8th. This will allow for more time to socialize and let the kids play. More information is posted on our web page at <http://galactosemiasupport.org>.

Another goal is to have an educational event in the fall of each year. Topics will include information on education concerns, latest research on galactosemia, and recipes or nutrition ideas.

Our first formal educational meeting was held in September 2006 in conjunction with the Dallas Children's Medical Center Genetic Clinic social. Ten families attended the event. Jo Beth Southard shared information she received at the 2006 PGC conference. There was also discussion about new educational laws and planning of our first social event. A roster was also developed so that GFSS could stay in touch with their members. If anyone has a topic for the meeting in September/October 2007 or knows of a speaker or is a speaker, please call Jo Beth Southard 817-832-7998.

GFSS will also help fund research for galactosemia. The first annual Sarah's Cure took place in October 2006. GFSS will be instrumental in this annual event. Also, Amanda Rawls organized a karaoke contest fundraiser in Tyler, TX in February 2007.

GFSS wants to support all families who have children with galactosemia. We will provide a packet of information to all genetic clinics in our region so that when a child is diagnosed with galactosemia, the parents will have access to an immediate support group.

We elected the following officers in September 2006:

President - Jo Beth Southard (817) 832-7998
Vice President – Mark Southard (817) 437-6147
Secretary – Amanda Rawls (903) 343-5220
Treasurer – Kellie Wilcox (903) 726-9446

Please feel free to contact any of us to join our group, with questions, or just for support. We would love to hear from you.

Please join us all in extending our best wishes to PGC's Secretary Nish Rao and her family as they welcome a new addition. Nish gave birth to a baby boy weighing 6 pounds and measuring 19 1/2 inches on April 8th, 2007. His name is Rohan Rao.

Congratulations to Nish and Gopi and new big brother Rahul.

PGC Logo Items Available

PGC logo items are 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 for pictures of these items.

Contact Michelle Fowler by email at president@galactosemia.org or by phone a 1-866-900-7421 to place an order.

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.

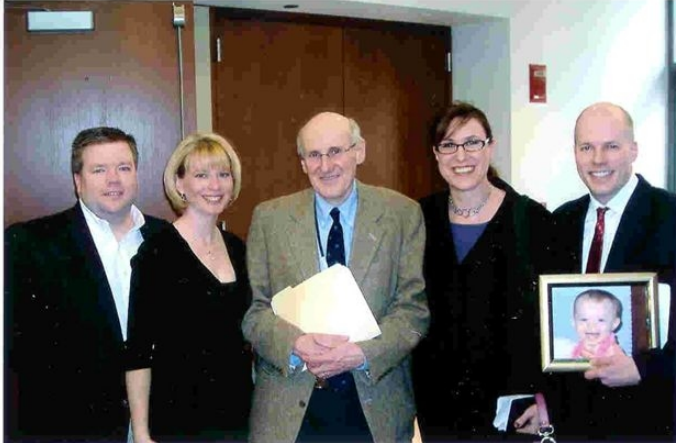
AAA Environmental, Inc. will supply us with NO COST postage paid recycling envelopes to send in our used printer cartridges and retired cell phones. We have recycling envelopes for you to use or give to family, friends, neighbors, etc., anywhere in the United States.

For more information or to obtain envelopes, please contact
Michelle Fowler at president@galactosemia.org.

Maureen Bell Speaks at Children's Hospital of Boston

Maureen Bell
PGC Vice President

On Wednesday March 14, 2007 I had the opportunity to speak to the Harvard Medical School community at the Children's Hospital of Boston about my life as a galactosemic.



(L-R) Bill Bell, Maureen Bell, Dr. Gerard Berry, Kristine Lydon, and Sean Lydon at the Grand Rounds Presentation at Children's Hospital of Boston on March 14th, 2007.

I was part of the "Grand Rounds" presentation given by Dr. Gerard Berry, who recently relocated to Boston to become the head of the metabolic division at Children's Hospital as well as to develop the "Center of Excellence" for galactosemia research.

I spoke for about ten minutes and told the medical community about my life as a galactosemic and the many challenges that I have overcome. The audience included Harvard Medical Doctors, interns, residents, dieticians and nurses anxious to hear both myself and Dr. Berry speak.

Also in attendance were my husband Bill, my mother Rose Lindenhofen, my uncle Fr. Francis O'Rourke, and Sean and Kristine Lydon who spoke after me and told the story of their daughter, Kyleigh who also has galactosemia. Kristine talked about how the metabolic clinic has changed so much for the better since Dr. Berry has arrived.

Dr. Berry presented the overall view of galactosemia to the medical community prior to my talk. He was warmly received by all in attendance.

Afterwards, Maureen and her family were given a tour of the hospital and also where Dr. Berry is working to open the Center of Excellence.

2008 Conference Update – Chicago Here We Come !!

We are pleased to announce a few more details about PGC's 2008 Conference.

As was announced in our Fall 2006 newsletter, Chicago will be the host city!

Chicago is a centrally-located and "easy-to-get-to" city. With plenty to see and do conveniently accessible by public transportation, it's a friendly place for families and children.

Our conference will be held on **Friday and Saturday, July 18th and 19th 2008** at the **Crowne Plaza Chicago O'Hare hotel**. The Crowne Plaza Chicago O'Hare is located in the heart of Rosemont at 5440 N. River Road, Rosemont, IL, about 20 minutes from downtown.

Attendees of our conference staying at the hotel will enjoy reduced room rates of approximately \$150.00/night, inclusive of taxes and will be offered complimentary shuttle service to and from the O'Hare Airport.

Details of the procedures for making reservations (either via the hotel's website or by telephone) will be available soon and will be announced on the PGC website, via e-mail and on the www.galactosemics.org message forum.

Your PGC Board of Directors and the wonderful conference committee volunteers are continuing to work on the details of this exciting event (e-mail conference@galactosemia.net to volunteer your time and talents!) . We will keep you posted as things progress. Please go ahead and mark your calendar now and make plans to join us for our next conference.

Support to Adults with Galactosemia

Sharon Howell

Chairperson, Support to Adults with Galactosemia Committee

I recently asked to volunteer on the committee to Support Adults with Galactosemia. I believe it will be a great opportunity to share my own experiences with others familiar with galactosemia. At age 47, I have experienced a life of managing my special diet, grieved because of infertility due to galactosemia, and most recently, had a hysterectomy. In addition to sharing my own experiences, I hope to learn more from others who must live with this condition.

Parents of Galactosemic Children (PGC) has given people like me the opportunity to share concerns, experiences, and help answer questions regarding galactosemia. To those persons with galactosemia, we have each had our own problems and complications due to this rare condition. Yet, thanks to PGC, we now share a bond that can never be broken. In the past, we may have felt isolated from others with galactosemia. This organization has brought many of us together for the first time. It is within this group where we may find some answers and support never before experienced.

I am one of the oldest females living with galactosemia. I have some problems, but it is unknown if these problems are galactosemia related. Most of my problems began when I learned that I would be unable to have children. I was 19 years old, and I felt that my world had been shattered. I had no one to talk to about infertility; at least no one who could understand how heart breaking that was for a young female.

Since attending the PGC conference in Philadelphia, in 2006, I have had the opportunity to correspond with others touched by galactosemia. My most precious letters and e-mails have been from women in their 30's and 40's who have experienced infertility first hand. I understand their pain, and I share their sorrow.

It is a mystery why I have not suffered some problems perhaps associated with galactosemia. For example, school was fun and sometimes easy for me. With the exception of math, I did not struggle during my pre-college academia. I enjoyed school. However, I have been in contact with others who have galactosemia, who have told me that school was or is very difficult.

College was different for me. About the time I was preparing to attend college, I was learning a difficult lesson about infertility. I was depressed most of my college years. Sadly, my heart was never into advanced education after learning about infertility.

My husband, Marc, and I are anxious to work with others interested in Supporting Adults with Galactosemia. However, in order to fully assist families and persons with galactosemia, we will need assistance from those who have experienced the struggles and difficulties that have so far, eluded me. We are open to ideas and suggestions on ways to improve the lives of those persons with galactosemia, and their families.

If you or someone you know is interested in assisting us, our e-mail address is newzjunkie@aol.com.

In Memoriam

It is with great sadness that we report the passing of a true pioneer in the field of clinical galactosemia research and a longtime friend to PGC. Dr. Stanton Segal passed away on April 16th, 2007 after a brief illness. He was described by a protégé as "*an ideal mentor ... nurturing, tireless, inventive.*" Dr. Segal was the longtime Chief of the Division of Clinical Metabolism and the Director of Division of Biochemical Development/ Molecular Disease at The Children's Hospital of Philadelphia (CHoP). He was also connected with the Division of Human Genetics and Molecular Biology at CHoP and the Dept. of Pediatrics, University of Pennsylvania School of Medicine in Philadelphia. He published more than 400 papers and received over \$20 Million in research funding during his 40+ year career.

Among the many honors he received over his career, Dr. Segal was the 1998 awardee of the Robert H. Herman Memorial Award. This award is given each year to a clinical investigator whose research work has contributed importantly to the advancement of clinical nutrition in areas particularly involving the biochemical and metabolic aspects of human nutrition. The entire PGC community extends our most sincere condolences to all who mourn the passing of this great man whose life work helped so many.

Lyndsey's Courageous Story

Achieve – Believe – Dream

Linda Leggio
PGC Founder

Lyndsey is Julie and Mike Meyer's third child – their first Mark (21) does not have Galactosemia, but their next two Bobby (20) and Lyndsey (17) both do. Watch for an article in the future about Bobby who has Asperger's Syndrome and the challenges he has overcome.

This story, however, is about Lyndsey. Three months prior to Julie getting pregnant with Lyndsey she went on the galactose/lactose-free diet and of course followed it throughout her pregnancy – four months into her pregnancy, an amnio confirmed that Lyndsey did in fact have Galactosemia. Back then, it was thought (or let's say "hoped") that if you followed the diet during your pregnancy that your Galactosemic child would be fine – we have learned through not only this family but others that this is not always the case.

When Lyndsey was about 4-years-old, Julie noticed that when they scolded her she would get "nervous" – shaky. She responds poorly to authority – this is one of those things that apparently is quite common among Galactosemics. They then noticed that she was having difficulty with fine motor movements, so she started occupational therapy. Tremors were still not mentioned or part of the equation. Then, when she was in 5th grade, Julie noticed that "the way she talked had changed, it sounded like she had a frog in her throat." At that time Lyndsey was diagnosed with vocal nodules and started speech therapy, which is when they were told that she also had a vocal tremor. Around the same time, it became apparent that she was having difficulty writing – "she held her pencil in a weird way," says Julie, "it was because she was compensating for the tremors and it was the only way she could write."

Like many other families whose kids have tremors they took her to neurologist after neurologist ... all of whom would say things like, "I've never seen anything like this before," or "I'm not quite sure what to make of this." And that's when they like other families began trying the various medications available for tremors – none of really which work – as many of us have learned. Many of us whose kids have tremors have compared notes over the years and have been continuously frustrated – Julie was no exception.

Lyndsey's tremors were continuing to get worse, which has been seen in many of the Galactosemics who have tremors. Finally, about a year ago, Julie got Lyndsey in to see a well-known pediatric neurologist at the University of Wisconsin who said, "You do know that whatever medication you put Lyndsey on her tremors will never go away and no medication will even make it 30% better." This was the first time Julie had ever been told this but of course it made sense since the meds they had tried over the years had not helped. The neurologist suggested deep brain stimulation also known as DBS – this was the third neurologist to suggest this. As the doctor put it, "It's obvious that the tremors have taken over Lyndsey's life." (*Note: DBS has been suggested to other families as well. Julie had never considered it as an option since Lyndsey was so young.*)

The kids at school were making fun of her calling her "earthquake," not to mention it was very difficult to understand her because of the vocal tremors. In addition, Julie and her husband had finally told their daughter that because of the premature ovarian failure that Lyndsey suffers from she will not be able to have children. (*Side Note: There are two women who have POV who have had children, so there is still hope, although very little.*) The tremors in her hands were so bad that her writing was unreadable, and because of her vocal tremors it was difficult to understand her. Already self-conscious, depression began to set in, Lyndsey stopped eating at school because the kids made fun of her, in fact she didn't want to go to school at all. She was losing weight and becoming more depressed by the day. Julie was becoming desperate, her daughter was on a downward spiral and she had to do SOMETHING!

At that point a team of doctors got together to determine whether or not she was a good candidate for DBS. Lyndsey was not only the first Galactosemic to have DBS surgery but also only the 21st child (under the age of 18) in the country to have this surgery. After about a week, the team reported that Lyndsey was eligible for the surgery. They would perform the surgery on the left-side of the brain, which would improve the right side of her body – her dominant side. They only do one side at a time, if Lyndsey chooses to do so in the future she can have the other side done but the risks increase.

She finally got an appointment with Dr. Erwin Montgomery, a neurologist who specializes in Parkinson's Disease and DBS. He identified seven areas of tremor in Lyndsey – tongue, mouth, vocal, torso, arms, shoulders, hands, and she also has the awkward gait. He also said that she had a combination of tremors (see the “More on Tremors” article). He confirmed what the other neurologists had said, that Lyndsey was a perfect candidate and could benefit from DBS.

Lyndsey finally felt hope, something she had lost. As Julie says, “When she found out DBS could get rid of her tremors she became a different person, it was full speed ahead.” Thus began the journey that would change Lyndsey's life. They then met the neurosurgeon who specializes in DBS surgery, Dr. Leland Albright. One of the physicians present during the surgery commented to Julie, “To see these two doctors [Montgomery and Albright] work together is awesome!” There was no question in Julie's mind that she had found the “best doctors for Lyndsey.” As a side note, Julie has worked in PR in a hospital for more than 20 years and in addition to the knowledge you absorb as a parent of two Galactosemic children, she also brings a medical background to the table.

This decision, however, was not Julie's or Mike's, it was Lyndsey's decision all the way. The doctors told Julie and Mike, “if you came in and told me that you wanted this surgery for your child we would tell you no, the child is the one who has to want it.” Lyndsey's feelings, “If you think your tremor is ruining your life then I would recommend it ... but you need to consider the risks and it is a long and hard recovery.”

Prior to the doctors considering the surgery, it is required not only by the insurance company (it was covered) but by the physicians – that she try any tremor medications that have not been tried before. Julie had a list and proof of the medications that Lyndsey had been on in the past, which of course did not work. They were then given a list of medications that had not been tried, which Julie took to her pharmacist who determined which ones did not contain lactose or galactose. There were two that Lyndsey would have to try. They tried each one for a clinical trial of two months each – and they didn't work – this came as no surprise.

What followed was a series of appointments, they were shown a video of the procedure. Julie, went armed with her notebook – she had been researching DBS for more than a year and had a list of more than 30 questions, which were answered. She also asked if it made a difference that Lyndsey has Galactosemia, “the doctors looked at me like I was crazy, they said, ‘Galactosemia has nothing to do with it (the surgery), a tremor is a tremor and that's what we're treating.’”

They were told that the risk was 2 out of 100 that Lyndsey could get an infection (of the brain), stroke, or even death. As Julie said, “I know all about risks ... one in 100,000 children get Galactosemia and I have two! The risk of 2 out of 100 did not sound good to me at all.” Mike, Lyndsey's father said, “This is my baby girl and I don't like the odds.” Lyndsey was upset by her parents' reactions – she still wanted to do it. After a lot of soul searching and more, Julie and Mike finally agreed –DBS was the only hope anyone could offer – and it would change the quality of Lyndsey's life. The decision was made.

February 9th : The Day That Would Change Lyndsey's Life

(Disclaimer: Lyndsey's mother told the writer the following, it is from a parent's notes and point of view, please excuse any discrepancies.)

It took almost 2 hours to fit the halo on Lyndsey's head, which is necessary to prevent her head from moving (she was fortunate, because she was a minor she did not have to have her head shaved). She was supposed to be in a twilight sleep but it didn't work so she was awake during the entire surgery which took 9 hours for the first part and 2 hours for the second part (when “pacemaker” is inserted). She was on a mild tranquilizer, which she received through her IV. Her parents were not allowed in – there were about four surgeons, two anesthesiologists, a programmer, radiologist and more in the room. The toughest part for Lyndsey was that they were working “behind” her and she could hear them talking but could not “see” anyone, which she expressed after the surgery.

The surgery involves placing an electrode about the size of a pencil but thinner with wires coming off of it into the brain. The wires are then “hooked” into specific points in the brain; these wires are also hooked up to the pacemaker, which is inserted just above the heart. The pacemaker is what will eventually give her control over the tremors. During the surgery, Lyndsey was asked to perform a variety of tasks, e.g., move your arm, your leg, etc. This is done over and over as the surgeons try to find the part of the brain that “controls” the tremors. As Julie explains it, “There's a conversation going on in the brain and the doctors understand it and Lyndsey's movements and “talking” direct them on where to go.” She was also asked numerous times to say, “During the British Constitution” and “Today

Lyndsey's Story (continued)

is a lovely day" ... Lyndsey who has a great sense of humor would say, "Today is NOT a lovely day."

After the Surgery

Immediately after the surgery, Lyndsey's tremors were completely gone. But the doctors warned that this was not going to be the outcome – apparently, the brain gets "shocked" during the surgery and as a result the tremors disappear completely – but that only lasts about 2 weeks, and that was the case with Lyndsey. The family gathered in her hospital room once she had recovered – the nurse brought in her dinner, which included a bowl of soup. For the first time, Lyndsey ate the soup – "It was like a party as we all watched, she was never able to eat soup, she just couldn't get the spoon to her mouth without spilling it all over. No more sippy cups, no more straws ... a whole new world was opening up for Lyndsey. Of course, she was still in the early stage and we knew that this wouldn't last but that moment was one we will never forget," says Julie.

As Lyndsey puts it, "The outcome was better than what it was before but we don't have 100% yet and if you think that it's going to be perfect like right after the surgery you will be disappointed; and it will take a lot to get there ... to 100% and it may never get there ... to 100%." It's been two months now and Lyndsey is at about 70% -- Julie said she'd be happy with 50% but the doctors say they will only settle for 100%. Her vocal tremors and handwriting have also improved.

Lyndsey now goes to a "programmer" who works with the neurologist. Basically, the pacemaker needs to be programmed, there are a series of thousands of equations and they just have to find the right one (or fine tune it) so that will control the various tremors. It's a tedious process as Lyndsey is asked to pick up a cup and put it to her mouth over and over while the programmer works to get the tremor under control. "It's amazing," says Julie, "they have the arm and hand under control but not the shoulder, so that's what they are working on now. Lyndsey can actually feel the tremor coming from her shoulder." The programming process will go on until they get it perfect at which point Lyndsey will be on her own.

Lyndsey says, "Programming takes a lot of time, months, and it is tiring on you and your brain, and you need to know that it may not fix all of the tremor but we don't know that for sure." She has a "remote control," which gives her the option of turning it on or off. Also when she goes to the airport she needs to tell security about her pacemaker and sometimes it can be "tripped off" so she needs to have the remote to turn it back "on."

After the surgery, 28-year-old Holly Harrison who has Galactosemia and tremors sent Lyndsey a bracelet that she wears all the time, it reads "Achieve – Believe – Dream." The card that Holly sent with it read, "Dare to dream whether big or small, believe in yourself and you can achieve it all." For Lyndsey, she believed and has achieved her dream, which is slowly coming true.

If you have any questions or would like further information, you can contact Lyndsey's mom Julie Meyer at jrmjmeyer6@gmail.com or 608-849-7974 (Wauwaukee, Wisconsin).

Summer Camps for Speech, Language, and Learning

In anticipation of the need for some parents to find appropriate summer camps for their children with apraxia, we have compiled a list of programs which may be appropriate for children with apraxia and related issues. We have only included camps that are holding sessions this summer. They are listed in alphabetical order on the PGC website. If you are interested, please contact the camp to find out if it is suitable for your child's particular needs.

Please note, inclusion of a camp in the listing below does not constitute endorsement by PGC. The listing of camps is for information purposes solely. Parents should carefully review camp materials, interview, and thoroughly investigate programs.

More on Tremors

Notes from a Visit with Dr. David Lynch of Philadelphia Children's Hospital

Linda Leggio

My son Adam Manis and Susan Fernstrom's daughter Holly Harrison both participated in Dr. Stanton Segal's PET/MRI research study in June 2005. One of the physicians who Dr. Segal is consulting with is neurologist Dr. David Lynch, who as a result has developed an interest in the neurological issues that some Galactosemics are dealing with.

Adam and Holly met with and were examined by Dr. Lynch the morning before the PGC Conference in July 2006. He quickly put both Holly and Adam at ease and took more than just a professional interest but a personal interest in their lives as well. Similar to other neurologists Holly and Adam have seen over the years, Dr. Lynch too, had never seen the variety of "symptoms" they display. The difference this time though was that this was the first time that both Susan and I felt that we finally got some "concrete information" about our children's tremors and more.

Basically, he said that they both have a combination of three types of tremors – Cerebellum Tremor, Essential Tremor, and Dystonic Tremor. Previous neurologists had mentioned the first two, but not the last, Dystonic Tremor, which Dr. Lynch felt was the most prevalent among the three.

He noticed that both Holly and Adam tilt their heads to the side, this is so slight that Susan and I have never noticed it – it can be a symptom of progressive neurological changes, such as those in which there's a disconnect from sensory input to motor output. He mentioned that he'd like to see pictures of the kids when there were younger to see if the "head tilt" was apparent and/or determine the age when it did become apparent. He also wants to see their MRI films from past years (these can easily be downloaded to a CD).

When we mentioned some of the symptoms we see such as Adam's rocking, or Adam and Holly's stiffening of one arm and holding it up – he explained that rather than necessarily being alarmed by these "behaviors" that may NOT be a sign of deterioration but that these behaviors could be "evolving" because of the way that their brains and bodies "learn" to cope with the neurological issues. This made sense to both Susan and me, because it means that our kids are not necessarily getting worse but that their bodies are compensating and evolving.

We discussed medications, deep brain stimulation (DBS) (see "Lyndsey's Story" in this newsletter)– which one of Holly's neurologists had mentioned to Susan, and Thalamotomy. The last two are invasive procedures, which are only recommended when all other treatments (medications) have failed.

Dr. Lynch discussed three medications, listed in the order of his recommendation:

- 1) BuSpar (Buspirone) – an antianxiety agent used primarily for the relief of mild to moderate anxiety and nervous tension. (Adam's pediatric neurologist had mentioned this once but we had never tried it because two of the side effects can be sleepiness and sometimes confusion.)
- 2) Baclofen (Lioresal) activates GABA type B receptors that are found in the spinal cord. As a result it is particularly useful in spasticity – which both Holly and Adam also display, i.e., the stiffening of the hand and holding it up
- 3) Artane (Trihexyphenidyl) – which is often used in conjunction with other drugs for the relief of symptoms of Parkinson's disease, which causes muscle tremor, stiffness, and weakness.

Dr. Lynch said that Holly and Adam do not have the classic Parkinsonian symptoms but that the treatments used for Parkinson's disease are *sometimes* also successful with tremors. On that note, Adam takes Sinemet, a Parkinson's medication. Adam went off of it for about 2 months last summer for the Segal research study (he could not be on any meds). I saw some worsening of the tremors but it was hard to tell because we were traveling and he was under stress, which always makes the tremors much, much worse. When I asked Adam if he wanted to go back on the Sinemet, he said yes, that he felt it made a difference.

Neither Susan nor I have made a decision about the medications at this time. BuSpar would be our first choice and as Dr. Lynch explained if they start displaying the side effects then it's easy to discontinue.

I asked Dr. Lynch to review this article for accuracy before submitting it to PGC, since I'm only a parent looking for answers.

Not all patients with galactosemia develop tremors, the current research shows that it's about 20% of the galactosemic population and there are varying degrees (like everything else with Galactosemia).

Dr. Lynch has agreed to "speak" with other families whose Galactosemic children have tremors and/or are showing signs of other neurological disorders. His e-mail is below. If there is any way that you can arrange a visit to Philadelphia to see Dr. Lynch, we encourage you to do so. The more Galactosemics that Dr. Lynch examines may eventually help with some answers.

Dr. David Lynch: lynch@pharm.med.upenn.edu

If your child has tremors, I have put together a "survey" that I would like to send you so that we as parents can begin to "compare" and work together with these physicians and with one another to help our children.

Linda Manis: LMScript1@aol.com or 561-862-0748

Susan Fernstrom: fourwillow@comcast.net or 978-388-2181

A Note to Families of Newly Diagnosed and Young Children with Galactosemia

I often write articles about the young adults with Galactosemia, the difficulties they have, and most often about the neurological issues – tremors. Whenever I write these, I cringe at the thought of you reading these and the fear and anxiety you must feel about the unknown. In PGC's efforts to address everyone, it is important that articles such as these are presented. But it does not, by any means, imply that your child is going to develop these problems.

In my experience since founding PGC more than 20 years ago and talking to probably more than a hundred families and numerous physicians, I have come away with my own impression that about 20% of the children are perfectly fine with no problems at all; 20% have a multitude of problems but are often able to function in society (this would include my son Adam, now age 23); and 60% in the middle who may have mild to moderate complications with areas such as speech, learning, or other such difficulties.

My advice to all parents has always been, "Go with your gut feeling," if you think there is a problem, act on it immediately, get your child evaluated, and seek answers from a professional. Talking to other families within the PGC circle is also important, as the families run the gamut.

And as a parent of a child who is in the 20% range with a multitude of problems, I can tell you that I do not know what life would be like if Adam were any different. Anyone who has ever met Adam has been instantly impacted by his warm, friendly demeanor and of course his smile. And although Adam does not talk much because of his speech delay, he does communicate on the computer and when he writes we realize there's a whole other person in there who is amazingly quite smart.

Linda Leggio

Making "GANES" for You!!

Spring 2007 Update

Announcements

Website: Check out our brand new website!! GANES is so appreciative of our volunteers who helped to make this happen! David Reid and his company gave our site a sleek, professional look! Patrick Fernstrom continues to host our site. We would also like to thank the Miele-Herndon family for letting us take over the galactosemia.com address. Check out our new site at www.galactosemia.com

Spring Gathering/Fundraiser Baseball Game: Take "GANES" out to the ball game! Get ready baseball fans - GANES is having an afternoon at the Camden Riversharks game, in Camden New Jersey (just over the bridge from Philadelphia), on Sunday June 17th. This is also Father's Day - and what better way celebrate than by taking the GANES' dads out to the game! This is a combination fund raiser and social event for us. We are looking for volunteers to sell game day tickets and we will make at least \$4 for every seat sold! There are 2 types of tickets available – for the game on June 17th and an undated voucher that can be used on a game of your choice between Sundays and Thursdays.

Please contact us if you would like to attend and if you are able to help sell tickets or if you have any questions.

Current Projects

Fundraising through the inkjet and laser toner recycling program:

This continues to be our main source of income. We have a few families in the GANES region who have begun to recycle in their area. It's simply a matter of word of mouth to local businesses, families, co-workers, neighbors, churches, etc. The collection point-person then boxes the cartridges up and they are mailed with postage paid labels to the recycling company. This trash-to-cash program has helped us to do all of our programs to date. If you are interested in being a point person in your area, please contact Cheryl Forsyth for information at cjanie1210@verizon.net

New baby gifts:

We'd like to help people to know that they are not alone, and that there is a network of support out there. To welcome new families to our community, we will send an insulated bag with GANES contact information and goodies for the baby. To date, 3 bags have been distributed.

Donations to support Dr. Segal's galactosemia lab:

Dr. Segal, from the Children's Hospital in Philadelphia, has been working in galactosemia research for over 50 years! He currently has some projects that are in jeopardy of stopping due to a decrease in funding support. To help him to help "us" we have promised to do what we can to fundraise to support his laboratory and research.

So far, we have sent \$2000 to Dr. Segal's lab, through targeted donations that GANES received.

To make a targeted donation to Dr. Segal's lab, please make out your check to GANES and send to us with "Dr. Segal" written in the memo line.

Future Projects

Educational and social gatherings:

We would like to offer both social gatherings and educational meetings through GANES. We need your ideas and volunteer commitments to make this happen! If you would like to host an event in your area, please contact us!

More fundraising ideas!

We are open to any and all ideas. GANES is a 501 (c)(3) nonprofit, which means that donations are tax deductible in accordance with IRS regulations. If you are able to host a fundraising campaign in your area please contact us!

Your ideas!

GANES is here to serve our regional galactosemia community. We are run by a few volunteers and we need your help to guide projects that you feel are important. We want to hear from you!

We hope that this update helped fill you in on our activities and sparks an interest to get more involved. Making GANES for all of us is our goal!

Sincerely,

Angie Forsyth, President
Jane VanZandt, Vice President
Maureen Bell, Secretary
Mike Denny, Treasurer

To Contact GANES:

toll-free:	1-877-795-4895
email:	galactosemia@ureach.com
website:	www.galactosemia.com
mail:	GANES, Inc. P.O. Box 479 Woodbury, NJ 08096

Generation Gs

Be on the lookout in the near future for Generation G on the Web ... Check PGC's website www.galactosemia.org and click on the link for Generation Gs. From there, you will be able to click on the BLOG and post messages, ask questions, post pictures, and meet other young adult Galactosemics.

As mentioned in the last PGC Newsletter, Generation Galactosemics (Generation Gs) formed last year at the PGC Conference in Philadelphia. There, Galactosemic Adults from the ages of 18 through 31 met and enjoyed a fun-filled two days meeting doctors and other Galactosemia professionals, they toured Philadelphia, and socialized A LOT.

The Generation G Coordinators are busy planning the GG Program for the PGC Conference in Chicago in July 2008 – Registration information will be with the PGC Conference Registration information. We're hoping to have even more people in Chicago – if we have enough people the big event will be a party complete with a DJ!

REMEMBER at this conference 16 and 17-year-olds will be able to join in the GG Program – you are also invited to join the GG Group by visiting our new Web site.

IT'S ALSO important to know that many Galactosemics have different difficulties – and some don't have any at all – that's what was so wonderful at last year's conference, people who didn't have problems helped those who did have difficulties – the one thing that each of them had in common though was that they all follow the same diet and they all have Galactosemia.

We hope to have get-togethers around the country during the years that there are not conferences – and as the GG Group grows, kids who are younger will have a group they know that they can join when they get older.

In the meantime, sign up on the new GG Web site and begin meeting other Galactosemics your age!

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. This bracelet features nine Swarovski colored crystals and a unique heart charm. (see PGC's website for picture). Order yours today!

NAME _____

ADDRESS _____

CITY _____ STATE _____ ZIP _____

# of bracelets _____	size 6 3/4"	@	\$ 30.00 ea.	_____
# of bracelets _____	size 7 1/4"	@	\$ 30.00 ea.	_____
# of bracelets _____	size 7 3/4"	@	\$ 30.00 ea.	_____

Shipping fee \$ 3.00 per item _____

TOTAL _____

Please make checks payable to:

PGC

P.O. Box 2401

Mandeville, LA 70470-2401

PGC's Operating Income and Expenses - 2006

1/1/2006 through 12/31/2006

Category Description	Checking General	Checking Research	OVERALL TOTAL
INCOME			
2006 Conference Income	\$23,172.00	\$0.00	\$23,172.00
Bank Fee Refund	\$203.00	\$1.00	\$204.00
Donation	\$23,285.34	\$126,462.00	\$149,747.34
Fundraising Income	\$4,509.09	\$0.00	\$4,509.09
Interest Inc-Interest Income	\$304.58	\$209.39	\$513.97
Memory Donation	\$885.00	\$220.00	\$1,105.00
Opening Balance 2006	\$63,959.91	\$0.00	\$63,959.91
TOTAL INCOME	\$116,318.92	\$126,892.39	\$243,211.31
EXPENSES			
2006 Conference Expenses	\$48,209.85	\$0.00	\$48,209.85
Accounting	\$974.00	\$0.00	\$974.00
Bank Charge	\$203.00	\$1.00	\$204.00
Bank Supply Fee	\$0.00	\$24.56	\$24.56
Finance Charge	\$52.49	\$0.00	\$52.49
Fundraising Expense	\$1,963.10	\$0.00	\$1,963.10
Licenses and Permits	\$316.86	\$0.00	\$316.86
Office Supplies	\$539.65	\$0.00	\$539.65
Postage and Delivery	\$377.66	\$0.00	\$377.66
Printing and Reproduction	\$3,325.06	\$0.00	\$3,325.06
Research Donations	\$0.00	\$106,360.00	\$106,360.00
Return Check	\$88.00	\$0.00	\$88.00
Telephone-Toll Free	\$224.57	\$0.00	\$224.57
Transfer to Research	\$3,697.00	\$0.00	\$3,697.00
Transfer Split donation	\$50.00	\$0.00	\$50.00
Travel, Business	\$1,061.33	\$0.00	\$1,061.33
TOTAL EXPENSES	\$61,082.57	\$106,385.56	\$167,468.13
OVERALL TOTAL	\$55,236.35	\$22,747.88	\$77,984.23

Comment about Financial Statement

This Annual Financial Statement was prepared at the end of PGC's Fiscal Year 2006. It serves as the basis for the accounting work that is being completed so that PGC can continue to fulfill IRS requirements. For the 1st. Qtr. of 2007, PGC has received a total of \$ 86,067.38 in donations (\$26,244.25 for the general fund and \$59,258.48 for research fund) and paid expenses of \$2,222.12. The balances as of April 1, 2007 are \$79,258.48 in the general fund and \$82,571.01 in the research fund.



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: board@galactosemia.org

Please remember to keep PGC in your 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**

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

Please note: you may need to adjust your spam filter settings appropriately in order to receive these emails