

OUR GALACTOSEMIA STORIES

2006



REESE'S STORY

Our son, Reece was born on January 31, 1959, weighing in at 7lbs, 13 oz. The doctors had run the bilirubin tests. My understanding was all babies have a bilirubin count when they are born which goes up a little then back down where it's supposed to be. Reece's bilirubin level was going up instead of going down. It his level reached 19 he would need to be a blood transfusion. If the bilirubin climbed to over 22 it would cause mental retardation. When Reece was 5 days old we took him to Iowa City and had his blood changed. He had a rare blood type.

After we returned home, he continued to deteriorate. He did not respond to his feeding and vomited most of them. Within 3 days, we returned to Iowa City. He was also jaundiced. This was the same reaction our first baby had to his feedings. He was born in 1953, was in the hospital 26 days, and died without a diagnosis having been established.

Reece was in the hospital for 2 weeks and still had not gained his birth weight back. However, the hospital thought he was all right and we took him home again. Within 2 days, he had the same response to feedings and we brought him back to Iowa City. Reece stayed in the hospital another 2 weeks and then back home we went. After a day or two @ home, Reece was acting the same way again. We were back to square one and back to Iowa City. They ran several tests again and then decided to run one last test; one that they rarely run. This test gave us a diagnosis of classic Galactosemia.

They began feeding him Gerber's Meat Base Formula. It agreed with him and he began to show a definite improvement. Reece was in the hospital another 2 weeks and when we came home this time his color was good and I felt confident he was improving.

The doctor explained to us how rare Galactosemia is and the complications the genetic disorder carries with it. We didn't know which one, one's, if any Reese might have. He was still 2 lb down from his birth weight and had some catching up to do. By one year old, Reese had pretty much caught up to where he should be and was a happy, healthy baby boy.

We didn't know we had any genetic disorders in our families. It was a shock to everyone. After our 1st baby died, we had a daughter and another son. After Reece we had another daughter. Tests proved the oldest daughter had both normal genes and both the next son and the last daughter proved to be carriers.

I am a writer, having published a small newspaper for over 11 years. I am in the process of writing a book about the many things encountered along the way in raising a child with Galactosemia. My purpose is not only to possibly be of help to others, but also to educate and enlighten the public. I hope to have it finished by the time of the PGC National Conference. I am very much looking forward to the conference.

Submitted by:
Millie in Iowa

Stephen's Story

Stephen, our third child, was born on January 12, 1964. He weighed 6 lbs and 9 ounces and was a beautiful baby. The first sign of a problem occurred on the fourth day of his life when he developed jaundice. A blood exchange was performed and a few days later we came home thinking everything was fine. However, I noticed he would not drink much of his formula and he cried a lot. Five days later on a check-up visit, Stephen's doctor found his liver was enlarged and he had lost almost a pound. Our pediatrician, Dr. Wm. Farley seemed to know right away that Stephen had galactosemia. He was immediately taken to Rex Hospital in Raleigh, North Carolina where he stayed for seven days. His formula was changed to Gerber's meat base and blood samples were sent to California to confirm the diagnosis. At that time, I believe California was the only place that could test for Galactosemia. Stephen gives blood every year here in North Carolina for the testing of newborn babies.

Stephen did not do well on Gerber's meat base formula. Dr. Farley called Duke Hospital in Durham, North Carolina and talked with Dr. James Sidbury, a metabolic specialist, who changed Stephen's formula to Nutramigen. Stephen began to thrive and gain weight. We saw Dr. Sidbury once a month, then every three months for five years. Dr. Sidbury was a very dear doctor who was an expert in his knowledge of Galactosemia. His knowledge and kindness helped calm me and give me hope.

After seeing Stephen for five years, Dr. Sidbury was asked to come to the National Institute of Health in Bethesda, Maryland. At his suggestion, we took Stephen to Dr. Henry Kirkman, a metabolic and genetic specialist at UNC Memorial Hospital in Chapel Hill, North Carolina. Dr. Kirkman was also a wonderful doctor with a kind and gentle spirit. He and Dr. Diane Frazier, a genetic counselor and dietitian, have been invaluable to us for thirty-five years.

They are so gracious and knowledgeable and even though Dr. Kirkman has retired, I can call him at home if I have an important question.

Stephen worked very hard all through school and graduated from Liberty University in Lynchburg, VA. He is now working in our family owned building and land development business, Dan C. Austin & Associates. He leases office and retail space in our shopping center and writes the most precise letters of anyone in our company.

As we all know, there are many trials growing up with Galactosemia. It is not easy and sometimes we don't have all of the answers. When we have difficulties we are not always sure it is related to Galactosemia. However, I do know this; we have always had wonderful doctors to turn to just at the right time.

They have been our shining lights in hard times. Dr. Stanton Segal at Children's Hospital of Philadelphia is certainly one of those shining lights for all of us who have children and young adults with Galactosemia. His lifetime goal has been to "find a way to treat patients so they don't have all the complications that happen even with a galactose-restricted diet" and ultimately to find a cure. He has the deep respect and admiration of all who know him. Stephen, my husband and I are so grateful to have had the opportunity to meet such a fine man and dedicated doctor.

Finally, one day about a year ago when I was praying for Stephen, I read a beautiful verse. I believe God gave it to me and I would love to share it with you. "Have I not commanded you? Be strong and of good courage, be not afraid, neither be thou dismayed for the Lord your God is with you wherever you go." Joshua 1:9

Whenever I don't feel strong, lack courage, feel fearful and doubt, there is our Lord telling us the opposite. He loves us and is with us always.

Submitted by:
Joanne
North Carolina

Collin's Story

On Monday, July 1st, 1996, we were blessed with an 8 lb, 4 oz. baby boy, our first child. After many hours of labor, Collin was delivered with the help of another doctor and forceps.

The time in the hospital was fairly normal and Mom and Collin went home on Wednesday. On Friday afternoon, July 5th, we received a phone call from our doctor asking us if Collin appeared jaundiced or lethargic. He told us a test came back indicating that Collin may have Galactosemia. At the time, I didn't know what galactosemia was and the doctor wanted to repeat the test.

On Monday, I took Collin in for his one-week checkup and was told that Collin did indeed have galactosemia, a metabolic disease that is manageable by diet. The doctor told me that I had to immediately stop nursing and start Collin on a soy formula. We had originally planned to see a plastic surgeon on Tuesday, July 9th to repair Collin's cleft lip, but instead we were on our way to the University of MN Hospital in Minneapolis to learn more about Galactosemia and have our son's blood level tested.

Yes, we were overwhelmed with the questions about our family history and genetics. We were told we could control our son's disease with diet and a "list of no ingredients" it was a little more reassuring. Little did we know about reading product labels, the processing of foods, Gal-1-Phosphate levels, etc.

I grew up in a large family and learned to cook and bake at an early age; therefore, preparing food at home was not an issue. We learned to substitute ingredients in our recipes...for example using 1 cup of dairy free milk and 1 Tbsp. of lemon juice in place of buttermilk has been a real lifesaver.

We have learned to allow extra time in the grocery store and have taught both of our children how to read labels and look for milk (and other forbidden ingredients) in the listed ingredients. We have a new hobby, going into new grocery stores or dollar stores looking for "new" foods that are milk free.

Collin was in the hospital several times the first 3 years for pneumonia, double pneumonia, RSV, croup, etc. He would be hospitalized for a week at a time. Now, Collin receives an annual flu shot and we haven't been in the hospital since. Collin is 9 and will be ten in July 2006. He is in the third grade. He enjoys math and reading (especially, if they are books about dinosaurs or animals).

He likes to go fishing, play sports (golf, soccer, basketball, baseball, bowling, etc.), watch movies and draw. Some of Collin's favorite foods include, pizza, chicken drummies, hamburgers, hot dogs, lasagna, green beans, beats, and rice.

Collins knows that there are many foods that he can't have and he asks (even Mom and Dad) if we are sure he can have them. We have learned to have snacks in car, at school, at church, or anywhere that Collin may be offered food so that there is always something available for him to eat. We often get calls from the school nurse or an aide checking ingredients, but Collins knows that he has to bring food home that he gets in school or give it to his sister. We always have baked goods (cookies, cupcakes, buns, etc.) or pizza in the freezer to have on hand for those last minute invitations or celebrations. My advice to new parents would be to give your child choices and keep a positive attitude. It makes it a lot easier not only on you as a parent, but also on your child.

Submitted by:

Barb

Minnesota

Greta's Story

Greta was born on November 14, 2004. She arrived by c-section a few days earlier than we were scheduled. For 3 $\frac{1}{2}$ yrs I had only one child at home and we were all eager to have our newest addition. I arrived home from the hospital on Wednesday after a three-day stay in the hospital.

Greta was a typical newborn. She slept most of the day and got up for a bottle once a night. She was on regular Enfamil formula; my only concern was that she was not a good eater. We really had to work to get Greta to take her bottle. On Thursday, I noticed that the whites of Greta's eyes began to look jaundiced, but family members told me not to worry. On Friday, I received a phone call from the Medical College of Virginia that the newborn screening for Galactosemia had shown up positive for Greta. The doctor had us take Greta to the pediatrician for an evaluation.

The pediatrician who examined Greta said that she looked okay, but switched us to a soy formula, sent us to the hospital to be safe. We were seen right away, but the doctors in the ER were not familiar with the disease. "Galactosemia" was something that I knew about since my niece has the "Duarte" form. She was so helpless and there was nothing we could do about it. They admitted Greta overnight for observation and we were released the next day.

Greta is now 18 months old and is very beautiful and bright. If I didn't tell you that Greta is affected with Galactosemia no one would ever know. Greta gets her blood work done every 3 months. She is talking and has over 26 words in her vocabulary.

On March 24, 2006 we had baby number 3. Amniocentesis confirmed that he was a carrier. We tested him because we didn't want any surprises. I was happy with the results. I am so happy to have the family that we now have and I do not dwell on Galactosemia. I have watched as Helga, my oldest and Greta play in the back yard together this spring and realize that we have the most perfect family. I look forward to all of the experiences that we will have and I take it one day at a time.

Submitted by:

Melissa

Virginia

Hannah's Story

Hannah was born on September 21, 2005 in State College, PA. She was born 3 weeks early due to low amniotic fluid levels and at birth was small (5lb, 2oz), but healthy. The day that she was discharged from the hospital, we received a phone call that her newborn screening testing had not been performed. We took her back and had the screening test performed. While at home with our beautiful new baby, we received a call from the pediatrician that one of her newborn screening had come back abnormal. This was four days later. Her total Galactose level was abnormally high, but her GALT activity was within normal ranges. We had never even heard of Galactosemia and the pediatrician assured us it was probably a false positive reading. We went to the hospital the next morning for additional blood tests. What a scary evening! I remember looking at Hannah and crying. I only wanted to hold her and know that she was going to grow up healthy.

The next morning, at one week old, we took her to the hospital lab for her blood work. It was a terrible experience of phlebotomists trying to get blood from our five-pound baby. We made it through the testing and visited her pediatrician to discuss switching to soy formula. We still thought this was a false positive reading and this switch was only a precaution. There was a mistake made at the lab and the Gal-1-P and galactitol levels were not performed. Hannah had to have another heel stick.

One week later, we received the good news that the second heel stick came back normal and Hannah did not have galactosemia - the total galactose and the GALT activity levels were normal. We were so excited - our worries were over. It was only a few days later that the pediatrician's number appeared again on the caller ID. I listened as Hannah's pediatrician explained that the results from her blood work done at the lab and sent to the Mayo Clinic had returned and her enzyme levels were low at 3.6, suggesting that she might have galactosemia. We needed to choose a metabolic specialist and set up an appointment as soon as possible. Hannah had presented so many conflicting test results we were nervous, but anxious to find an answer.

The roller coaster we were riding just would not stop. We hopped in the car two days later and drove three hours to the Children's Hospital of Pittsburgh to meet with the metabolic nurse practitioner and genetic counselor. We had confirmatory testing done that day and two days later, through DNA testing, finally had an answer - Duarte Galactosemia.

Since Hannah's diagnosis, she is growing and developing normally. Her Gal-1-P levels have decreased to zero and her galactitol level has fluctuated within the normal ranges. Her enzyme activity is still low (around 2.2) and even though her DNA says Duarte Galactosemia, she is being treated as a classical patient because of the low enzyme activity.

Submitted by:
Amy, Pennsylvania

Claire and Allison's Story

Our fraternal twin daughters, Claire and Allison were born on February 2, 1999. Claire has galactosemia; Allison does not, but is a carrier. Claire's galactosemia was not diagnosed until 2 and $\frac{1}{2}$ weeks after she was born.

Claire's galactosemia was not detected by the heel prick test done at the hospital after she was born. We still don't know why the test was flawed.

Claire appeared to be mildly jaundiced, but otherwise fine when she came home from the hospital. She continued to appear to be fine for about two weeks after birth. Fifteen days after birth, she began spitting up much of her milk and was examined by her pediatrician. The cause of the problem was not determined. The spitting up continued and worsened and she was admitted to the hospital 17 days after birth. She was losing weight and was put on intravenous feeding. Despite many examinations and tests in the hospital her condition was not immediately diagnosed.

Claire was finally diagnosed in the middle of the second night that she was in the hospital (day 19) by a nurse who was trying to figure out why she received a false positive reading on a glucometer (blood glucose) machine. The manual for the glucometer stated a false positive reading might be obtained for patients who have galactosemia. The nurse suspected that Claire might have galactosemia. Claire's formula was changed to soy and Claire's condition began to improve. Additional blood tests confirmed the diagnosis of Galactosemia.

As an aside, we have thanked the nurse who diagnosed Claire's condition many times. Every year we send a Christmas card with a picture of Claire and Allison to the nurses at the pediatric ward in the hospital.

Submitted by:
Heide
Wisconsin

Christin's Story

Our hometown is Brigantine, New Jersey. Christin was born in Atlantic City, New Jersey on March 15, 1978. We took our beautiful daughter home after two days in the hospital. Everyone marveled at her gorgeous skin tone. Needless to say, that changed within days. Since I was breastfeeding, it didn't take long for the symptoms to show up (i.e. vomiting, lethargy, and jaundice.) She was admitted back into the hospital at a week old and had to undergo a full body transfusion during the two-week hospital stay. The physicians encouraged me to continue to breast feed, but I was so nervous at having a sick baby that I stopped shortly after getting her home after the hospital stay. We put her on Similac Regular Formula and of course, that resulted in the symptoms returning. Her Pediatrician kept performing one blood test after another, but could not figure out what the problem was. At one point Christin became so dehydrated we had to schedule family members around a 24-hour period to rehydrate her with Pedialite. I remember going into our bedroom during that time and placing a pillow over my head so my family wouldn't hear me crying. I was as distraught as many of you parents must have felt at one time or another.

Finally after six weeks of going back and forth to the hospital, and with numerous blood tests leaving her poor feet in a constant state of bruises; we asked that she be admitted back into the hospital. I knew I was losing her. She was so frail. She had another blood transfusion and was then transferred to St. Christopher's Hospital in Philadelphia, Pa. Christin was born at 7 lbs. 6 ozs. At six weeks young she was 6 lbs. 9 ozs. St. Christopher's began the family medical history, and you can imagine some of the genetic diseases they asked about. On the third day, she had an eye exam, which presented the beginning stages of cataracts. This led them to Galactosemia, but Christin could not be tested because of the recent blood transfusions. Low and behold - Christin's father and I were both carriers! We were relieved we finally had a diagnosis, but now what? I was sent home with a case of Isomil, and little else. It was now time to do the research for ourselves!

This is where my hero, my Mother stepped in. As a Registered Nurse (RN) she had access to medical information, but in those days there wasn't much and what information we found wasn't very promising. As for researching food ingredients, Mom wrote to as many food companies as possible to find out what was in their products. I still have some of those letters along with their responses.

We continued our follow-up visits at St. Christopher's until Christin was 4 years old. After a discouraging and frustrating visit with the specialist, we decided it was time to find someone who knew more about Galactosemia. It was then that Mom found Dr. Berry at Children's Hospital. I will never forget our first visit with him. Dr. Berry sat for 2 $\frac{1}{2}$ hours answering our questions, no matter what they were. We knew we had come to the right place.

Even now, I tear up thinking about how kind Dr. Berry was and the relief I felt that someone finally could provide me with the best information there was on Galactosemia.

It was Dr. Berry that connected us with other families in the tri-state area who had children with Galactosemia. This was pre-PGC and the lady who founded PGC was among that first group. Her babies were still in diapers then, and now one served as the 2nd VP of PGC and the other is an amazing big-hearted adult with Galactosemia. Linda Manis and I still remain friends after all these years.

As a parent of a child with Galactosemia, I've experienced the problems that go with maintaining the diet; sending Christin off to school when I wanted to hover over her as a protective shield; making sure she got the right education to accommodate her learning disabilities; and as she got older, wondering if she would be able to have children of her own. I had an amazing support group in my family and the Brigantine community. And, when we moved to Florida (Christin was 9 at the time) the diet was easier to maintain because of the abundance of Kosher supermarkets and Whole Foods became my favorite store. Still is.

We now live in Las Vegas. Christin is now 28 years old. She works at a local Hospital in Las Vegas in the medical records office as a scanner, taking patient charts and scanning them into a computer program. She has been doing this for over two years and has decided to go back to school to become a Medical Assistant. She was accepted into the program and will begin early September. This will certainly be a challenge for her, but she has support of her family and the school to help her succeed.

I am extremely proud of my daughter on so many levels. For all my worrying, she has become an amazing, loving, beautiful young woman and my best friend.

Submitted by:

Paula

Nevada

Sean's Story

Like some newborns, Sean became slightly jaundiced after he was born. So, after coming home, we took him back to the hospital for daily bilirubin checks. When the levels kept going up, Sean was readmitted to the intensive care nursery for treatment. His jaundice required more than phototherapy or "light" treatment. On day five of life, Sean was diagnosed with this rare metabolic disorder through a stat newborn screening. He was no longer allowed to nurse and was given a soy-based formula. The disorder had already affected his liver and made him a high risk for infection. He stayed in the hospital for 11 days to receive a plasma transfusion, antibiotics, and light treatment. Sean was fortunate to be diagnosed early and although we don't know if he will have complications in the future, today he is an active, happy, growing pre-schooled.

He is very outgoing and social and loves to be around people. He adores his big brother, our 8-year-old son Ryan, and wants to do everything he is doing. Sean goes to a morning preschool four days per week where his teachers are aware of his Galactosemia and are diligent about the food Sean has access to. He loves to draw and practice his letters and numbers. He takes swim lessons at our local gym. Basically, he is a typical four-year old, except for his restricted diet.

Galactosemia really affected our lives and was terrifying as the parents of an affected newborn. We felt strongly that we wanted to "do something," and create something positive out of our situation. So, in February 2004, we founded our own 501(c) (3) nonprofit organization called GANES, The Galactosemia Association of the North Eastern States. Because galactosemia is so rare, we serve 13 states in the northeast to reach a larger number of individuals and families, and we work closely with the national organization of PGC, Parents of Galactosemic Children.

Our Mission: To create and strengthen networking of individuals and families affected by galactosemia along with health care providers, and to support education and research.

Submitted by:
Angie
New Jersey

James's Story

James was born February 21, 2006 at MUMC Medical Center in Hamilton ON, Canada.

James was delivered by C-Section and by all accounts was a healthy 8lb 3 oz. baby at birth. We were in the hospital for 4 days after delivery during which time James seemed to be breast-feeding with no problems. Two days after we came home, he was throwing up everything he ate; it was bright yellow and this concerned us so we brought him back to our family doctor and then to the hospital.

When we got to the hospital James was diagnosed with a bladder infection. They thought this was the cause of his vomiting and put him on a 14-day course of antibiotics and IV fluids. As a precaution, they also asked me to go on a milk protein-free diet, since one of his violent vomits was after I had ingested a large amount of dairy products.

Several Gastro-Intestinal tests later, they determined he also had reflux and put him on another medication. They did a spinal tap, several blood tests, and ultrasounds of his head, liver, kidneys, and heart, all of which appeared normal.

A month after being in the hospital, James was still unable to hold down his food and his weight had dropped below 6 lbs. The doctors were stumped and my husband and I were getting very anxious. I probably should mention that they do not screen for Galactosemia at birth in Canada (that is changing as of next year).

The doctors began to think that Cystic Fibrosis was a strong possibility. In the mean time, James had been on several types of elemental formulas (progestomil, nutramagin, alementum to name a few); but none worked except for isomil. They decided to put him on a feeding tube using my milk protein free breast milk and isomil as food. He still continued to vomit, but the weight loss slowed down.

On the fourth day of the feeding tube, James aspirated (swallowed fluid into his lungs). Upon ultrasound/x-ray examination they found James had fluid in his lungs and fluid (ascities) around the organs in his abdomen. This was taken as a sign of malnutrition. Because of the fluid, he developed two hernias and his testicles became painfully swollen. The doctors decided to operate the next day.

After the operation, things took a drastic turn for the worse. His incisions would not stop bleeding and when tested for his liver function they found his hemoglobin, coagulation factors

and albumin levels were severely "out of wack". Within two days he was septic and ended up in ICU - his liver was failing.

We were greeted by a social worker and told to stay at the hospital over night. They even provided us with a room that belonged to a resident doctor. We were cautioned that he may be on a ventilator by morning and told to be prepared for him to go either way. Seven blood products were transfused and within hours, thankfully, he began to improve.

We were moved into a step-down ICU where our nutritionist recommended that we try another type of formula called Neocate. It was a "god" send. In the mean time, more doctors were brought in including infectious diseases (he had a 2nd bladder infection and an e-coli infection), metabolic, genetics, gastro-intestinal, cardiology, and three more pediatricians. In the end, metabolic made the initial diagnosis of galactosemia and genetics confirmed that it was a heterozygous allele mutation responsible for the galactosemia.

We were in the hospital for another two weeks while the neocate "kicked". He had to receive albumin continually and lasix treatments as well as oxygen at night, but the ascites was going down and his weight was coming up!!!!

On April 19th, James finally came home and we couldn't have been happier. Although the experience was quite scary, especially as first time parents, the love and support we received from so many of our friends and family left us with some fond memories.

James is doing quite well these days and is developing at the normal rate. He has a great personality and we are so thankful for everything everyone did.

We look forward to reading other stories and learning more ourselves.

Submitted by:
Janice
Canada

Nicholas's Story

Nicholas was born April 20, 1993 at Credit Valley Hospital in Mississauga, Ontario, Canada. He was born a healthy baby. However, after I nursed him, he became jaundiced. His bilirubin was very high so he was put in an isolet under lights to clear up his jaundice. He would get a little better so I was able to nurse again. His bilirubin rose to an abnormal level and he became dehydrated. He needed an IV that was put in his head. We were not prepared at all for that. The doctors did not know what was wrong with him and we were doing all kinds of tests.

Finally, a brilliant doctor named James McKinnon, discovered that Nicholas might have Galactosemia. After tests confirmed this, he was put on soya formula and started to thrive. It took 8 days to confirm this because hospitals in Canada did not check for Galactosemia during newborn screening.

Nicholas was sent to the Hospital for Sick Children in Toronto for observation. A genetic/metabolic doctor specializing in Galactosemia examined Nicholas. Her name is Dr. Annette Feigenbaum and she helped us immensely through the whole genetic counseling process as well as giving us information about Galactosemia and informing us about PGC. After 5 days at the hospital, he went back to the other hospital for a few days and was able to finally come home. It was Mother's Day and I was jubilant to finally bring my healthy baby home. Nicholas had some liver damage, but an infant's liver repairs itself and his did.

He was checked by a pediatrician regularly and continued to thrive on the soya formula, Prosobee. The whole experience was very frightening, but we were thankful that his genetic condition could be controlled through diet. He developed at a normal rate and began to gain weight and thrive slowly. Today, he is a healthy 13-year-old boy who is musically talented, athletic and has a great sense of humor. We are very proud of him because we know that he is determined to excel in his talents and does not let health problems get in his way.

Submitted by:

Teresa

Canada

Marissa's Story

Marissa was born in August 1995 in Germantown, Maryland. When she was born, we didn't think she was anything but normal. After she was about 5 days old, she began to vomit profusely and started to lose weight every day. We put her back into the hospital at 7 days old and after two more horrifying days in which we kept feeding her breast milk (doctor's recommendation) she was finally diagnosed with Galactosemia. The nurses and doctors at the hospital knew nothing about this disease and told us she would probably die. We had her baptized in the hospital and hoped for the best. After researching the disease ourselves, we learned that, although fatal if not controlled, it was not the death sentence handed down by her doctors at the time. Her newborn screening, for some reason, did not come back quickly and the doctors had to call to find out the results. They made that call after we had received her diagnosis from the lab at the Mayo Clinic. We immediately started seeing a Genetics specialist at Georgetown University Hospital. There they told us the heartbreaking news about ovarian failure and what we might expect down the road.

We were able to identify Marissa's disease early enough that she has not suffered any long-term physical health problems. She had small cataracts and a slightly enlarged liver, but these were soon corrected by restricting her diet.

Marissa has problems with long-term memory and struggles with math at school. She has an IEP and has had one since she was five.

As everyone here knows, living with a child who has a disability is difficult and we take it one day at a time. School is a struggle for her and trying to educate the schools about her condition is a hard struggle.

Thankfully, as with many galactosemics, she shows no outward signs of the disease and you would never know anything was wrong by looking at her. We have been blessed that she is doing so well physically and that she handles her condition so well.

We now live in North Carolina and the wonderful genetics team at the University of North Carolina Hospital at Chapel Hill sees her.

Submitted by:
Janet
North Carolina

Hannah, Jaida, & Logan's Story

Our galactosemia story began in September of 1998. I had just delivered two beautiful twin girls. They were 33 weeks along and weighed 4lb 1oz and 4lb 3oz. Hannah was born first by c-section due to presenting in a breech position and then two minutes later, Jaida was born. Since they were premature, they were transferred to a hospital that had a neonatal intensive care unit. Both girls were doing fairly well in the beginning; they needed a little help with their breathing, maintaining body temperature, and increasing weight gain. They had IV glucose for the first 2 or 3 days (which we were grateful for now) and then later, given breast milk through a tube down their throats. Occasionally, the nurses would let me bottle-feed the girls, but it would cause them to burn a lot of calories.

Soon after, they both developed jaundice and were placed under the lights. They looked so cute with their sunglasses on. Jaida soon was taken off of the lights. She started to thrive and gain weight. Hannah started to deteriorate. We didn't know at the time what was wrong with her. It was so sad to see her fighting for her life. She was so small and fragile.

Soon after, Hannah fell ill. The newborn screening came back with the results that she had galactosemia. The doctors reassured me that all they needed to do was to take her off of breast milk and placed on soy formula and she would get better. She got worse before she got better. Her blood was now septic and she got a blood transfusion. She had surgery to place a tube in a main artery of her neck in case the doctors needed to give her medication quickly. She became very anemic. The tubes running out of her body would leave little indentions on her swollen arms and legs.

Two weeks after their birth, I was able to bring Jaida home. I was still nursing her. She was doing great and gained a lot of weight. Although, I was thrilled to have Jaida home, it was tough to take care of her and have Hannah, who was still very ill, still in the hospital. I would hold Hannah as much as they would let me and pray for her. One long month later, I was able to bring Hannah home.

Hannah's development has been somewhat behind as she grew. She had a music and development specialist as an infant and toddler. In preschool, she had some OT work and speech therapy. She just finished 1st grade and she is a little behind in her class. Hannah needs a little more time to understand things, but she gets them because of the incredible amount of effort she gives. She is reading now and able to do addition and subtraction facts. She is the hardest worker of all the kids. She is very motivated and will challenge herself. She is a wonderful, big-hearted little girl.

Hannah is not the only person in our family with galactosemia. In January of 2003, we gave birth to Logan. We knew our chances of having another child with galactosemia and were very careful during pregnancy and after to restrict any products containing lactose or galactose. He is doing really well. He was somewhat delayed in speech early on. He had a music and development specialist as well who focused on speech with Logan. He has progressed really well and now has no problems. Logan is three years old and starting preschool this fall. He is very special, loving, and a blast to be around. He always has a smile on his face!!

Submitted by:

Kara

California

Landon's Story

After having our first of three children in November of 1995, Tori Nicalett; Landon was born on October 13, 1999 weighing 8 lb, 11 oz. Landon appeared to be extremely healthy and alert at birth. I tried to breastfeed Landon, but he would have nothing of (thank goodness). Landon was placed on a soy-based formula due to continual spitting up, slight jaundice and just an overall unhappy baby. I was told to pump my breast milk and store it for later use since breast milk would eventually be best for my child when he was better.

Landon continued on a downhill slide each day. We went in for our "normal check-up" at 1 week old. The nurse put Landon in a room and we waited for the doctor to arrive. The long wait led me to look inside Landon's file, which was left in the room with us. I discovered that his newborn screening paperwork (thanks to the State of Alabama) showed positive for Galactosemia.

When the doctor finally came into the room, I questioned him as to what that word was and what it meant. He simply told me not to worry about the result; it was probably a false result and even if it is an "accurate" positive result, it was so rare and "nothing" needed to be done for Landon at this point. He said he was disappointed that I had not been giving Landon the breast milk and felt he would show improvement in his condition if I would start breast feeding him. He didn't know what was wrong with Landon, but he was SURE it wasn't Galactosemia.

Feeling very confused about the issue, thanks to my motherly instinct, I searched my diaper bag for a pen. Unable to find one, I had a strong feeling that I needed to find some way to write down this strange word. I took a q-tip, dipped in iodine solution and painted the word Galactosemia on the paper Landon had been lying on for his checkup. I tore the paper off and raced home to look up more information on the Internet. The first site I found was "Tyler for Life Foundation". I was completely overwhelmed by the information and the more I searched, the more overwhelmed I became. I immediately sought out a 2nd opinion for Landon and demanded repeat newborn screening be done. A STAT newborn screening was ordered and we quickly found out that he did indeed have this rare metabolic disorder.

Overall, we have to thank God. First, for our son being a thriving Galactosemic boy. 2nd, we thank him for leading me to be so determined in the doctors office to learn more about Galactosemia. We strongly feel that if God had not taken control of that particular point in my life, Landon would not be with us today. I could not imagine life without his laughter, country, southern boy accent, little mishaps and tears.

As the parents of a *Galactosemic* child, it is our goal to continually broaden my knowledge of the disorder to provide the best care for our child, as well as to increase awareness of *Galactosemia*.

Since the birth of Landon, in October of 2001, we had our third child, Tela Nicole who is a carrier. I did follow a strict *Galactosemic* diet during my pregnancy just in case. I found out immediately by cord blood that she was a carrier. Today, Landon is very much like any other 6 year old in his accomplishments and activities. We have no way of knowing if he will have complications in the future or how serious they could possibly be. He is only different in what he eats and drinks.

We can only say to parents of *Galactosemic* children, you are special and so is your child.

Submitted by:
Michelle
Mississippi

Kyle's Story

At one week of age, Kyle was throwing up and lethargic. That same day, I received a call from the pediatrician that one of the newborn screening tests came back positive and I needed to stop breast feeding and put him on soy formula. I was informed to take him to the metabolic clinic at Children's Hospital to have the test repeated. I had to wait a long 2 years to find out the final results. I was informed it was positive from a physician who was standing in for the Children's Metabolic Staff. His information about a child with Galactosemia was not very positive. I was devastated and cried all evening. Looking back, we were very lucky. Kyle was never hospitalized or ill in his infancy.

My son is now 15 years old and is a healthy well-adjusted teenager. It was not always a happy story raising a child with a metabolic disorder. We were lucky over the years that he never snuck any food. He was not happy many times when he couldn't have something, but he never cheated. Well, one time in preschool one of the moms offered Kyle a piece of chocolate cake; he did not turn it down, but freely told his father of his indiscretion. I won't go into details of what happened next at the preschool.

I would be happy to share any information to parents of how we have dealt with many situations over the past 15 years.

Submitted by:

Sherri

Colorado

Kristen's Story

My husband Dan and I have just come from Kristen's Middle School and watched her pick out her first high school courses. Her guidance counselor suggested all regent courses. She has been on High Honors eight out the last ten marking periods at Emmet Belknap Middle School in Lockport, New York. Thirteen years ago, we were unsure of where her journey would take us and we can remember like it was yesterday the day we were told Kristen had Galactosemia.

Kristen's first visit to the pediatrician was when she was 8 days old. He immediately expressed concerns because Kristen had lost nearly 2 lbs. He asked if she ever vomited and we said, "this baby is so good, she never even spits up and she is the perfect burper too!" When asked if she had loose stools, we told him her stools were already formed. He was still very concerned and made us another appointment in 1 week.

When we got home we had several urgent messages to call Dr. Baier immediately. Dan called the doctor right away and that's when our world turned upside down. Dr. Baier explained that Albany had called him that afternoon shortly after we left his office. We were told that Kristen's newborn screening test had tested positive for Galactosemia. I had saved the pamphlet we received from the hospital and retrieved it and read it. As soon as I read it, the tears began to fall.

We were instructed to call Buffalo Children's Hospital immediately and a specialist would be waiting to hear from us. We called and the specialist said, "you must be Mr. Holler", that's when we knew it was serious. We were told to take Kristen to Children's hospital within the hour and not to feed her any more milk. They said she could have a bottle of sugar water instead.

Arriving at the hospital, we entered a world we were not prepared for. It took awhile for the staff to contact the specialist since she was from the metabolic clinic and not a regular in the ER. Kristen was lethargic and we were unable to wake her up. We had to try to get Kristen to take a bottle of soy formula and were having a great deal of difficulty. The staff took a lot of blood from Kristen and us and ran other tests. Kristen was admitted to the hospital.

The next day, specialists came in and examined Kristen's eyes. No cataracts. They had such a hard time drawing Kristen's blood that Kristen cried so hard she vomited. Her throwing up led to projectile vomiting. Every time we got her to respond enough to make a bottle, minutes after it went down, it came right back up.

The next morning, I noticed Kristen's wrists were bleeding under her band-aids. The nurses thought this was impossible, since it was greater than 24 hrs since she had had a blood draw. But sure enough, she was bleeding. Kristen needed a blood transfusion right away and then was transferred to the ICU (I could no longer stay with her).

It was after dinner before I was allowed to see Kristen again. When we were allowed to see her, we had to don gowns and scrub up. The first time I saw her, my legs became wobbly and I didn't think I would be able to stand. She had nothing on but a diaper and she was on a square bed under lights. She was being fed through her belly button. She had lines and monitors hooked up to various parts of her body that were connected to machines that beeped behind her on the wall. She was also receiving nutrition through her IV. Kristen was still comatose, but she was sobbing in her sleep.

On day 5 in the hospital, Kristen began to improve. She was moved out of ICU and we were allowed to try bottle-feeding her again. At first, she would still projectile vomit, but later on in the day she started drinking and keeping it down, and finally thriving!!! Kristen came home on the 7th day when she was 15 days old.

One of the things we are most proud of is, Kristen goes to Strong Memorial Hospital in Rochester and speaks in front of learning doctors. Kristen always makes them applaud. We were just invited back for our 3rd year.

We know another journey we are going to face is possible premature ovarian failure. We have always been honest with her about this. Kristen says if she can't have children she will adopt and love that child as her own.

Kristen would like to speak at a Galactosemia Conference someday and we hope to come to meet other families so similar to ours this year at the conference.

Submitted by:
Beth
New York

Matt's Story

Matt was born weighing 12 $\frac{1}{2}$ lb and 24 inches long. I immediately knew something was terribly wrong. By the time he was 12 hours old, the doctor agreed with me. There was projectile vomiting, general malaise and jaundice. It was all downhill from there. He was malnourished, had cataracts, his blood count was 7 and had cirrhosis of the liver. I went home and Matt went to the NICU. They continued to try and diagnose him for the next 3 weeks. He lost a lot of weight and by 3 weeks was only 9 $\frac{1}{2}$ lbs. The specialists at the Children's Hospital were baffled. Our family physician was obsessed with the case.

When Matt was 3 weeks old, the hospital said we could take Matt home. We were stunned! He was still so ill!! We gathered his "coming home" outfit and went to get him. The head of pediatrics literally told us that we should take him home because he would probably die. He was near comatose and still; we took him home. They advised us to give him only formula, but of course, each time I did, he vomited. I started to give him baby fruit and anything else he could keep down. He even refused his formula. As he declined even more, the family physician called us each day for an update. He said he trusted my judgment and my observations could be valuable.

Finally, at 2 months, he called us one night at midnight and asked if we would take Matt for yet one more blood test. It seems he remembered something he had learned at a summer seminar. We went and the test was sent to USC and the diagnosis was *Classic Galactosemia*. This was 1974 and there was very little known about the management of the disease.

The doctor gave us his medical books and we began to study and worked on a diet to get Matt strong. It was a long road, but he began to thrive and get healthy.

School years were very difficult. I just couldn't get the educators to understand his problem. I gave them literature to read (which they obviously did not), but most of them thought he was just stubborn. Mostly, we worked tirelessly every night to help him reach his potential. We also used tutors.

Matt has grown into a wonderful young man, caring and personable. He is employed at Goodwill Industries, has received many awards from them for outstanding performance and was their featured employee for the year of 2005. We are extremely proud of him.

Submitted by:

Janet

Ohio

Gavin August's Story

Gavin August was born on April 19th in Fair Oaks, VA. He was a beautiful bouncing baby boy and I was so excited to finally be a mom. I just wanted to take our sweet little baby boy home. We were fortunate enough to have had a normal delivery and recovery so we were able to go home that Thursday. We really enjoyed our time as a family. It was great getting to know this little person and learning how to survive baby boot camp. In retrospect, I can clearly see that our little boy was not right. He was extremely lethargic and orange from jaundice. Since I was a new mom, these are things I didn't notice or know enough to worry about.

Saturday morning, I received the worst phone call of my life. My mom came into my room, woke me up from a much-needed nap, and handed me the phone. The call was from a Pediatric Doctor from the University of Virginia where all of the PKU screenings are sent. He informed me that Gavin's PKU screen came back positive for Classic Galactosemia and that I had to stop breast-feeding immediately. I had no idea that my son had even had a PKU screening done and I knew nothing about Galactosemia.

The doctor had attempted to contact my local pediatrician prior to calling me, but it was Saturday and he was unsuccessful. He was upset that he had to call me directly, but he was very kind and very worried about Gavin. This doctor felt it was important that Gavin be seen today, Saturday, rather than wait until Monday when the Pediatrician's office would be open. I truly feel this Doctor went above and beyond for our son and his future. His Classic Galactosemia was diagnosed within 4 days of his birth.

Once we got to the hospital, no one had ever treated Galactosemia. They also felt that Gavin was jaundiced and needed to go under the light. We were sent to another hospital where Gavin was under the light for 2 days.

He was given an I.V. and I began to bottle feed him with a soy formula. He vomited the entire hospital stay and also had an irregular, slow heart rate.

Once things settled down we were released with instructions to return to the hospital if Gavin began vomiting again. Gavin continued to vomit and we were back at the hospital in a couple of hours. Gavin had been placed on Similac Soy and it was decided to try something else. He was placed on Nestle Good Start Soy Formula and was able to tolerate that. Gavin lost 2 lbs. during this ordeal, but has made up for it now.

Gavin has been on Nestle Good Start Soy formula for the past year. Our insurance company even covered 75% of the cost of the formula, which was a huge help. He has had 2 blood tests so far and we have been referred to a Genetics Specialist as well as a nutritionist at Children's Hospital in Washington, D.C. Our doctors have really kept us informed and have helped my family through a difficult beginning. They are so positive about his future because they feel his Galactosemia was caught very early.

Gavin just turned one in April and my husband and I are enjoying every minute with him. Things are a bit more difficult now that Gavin has started solid foods so I have turned to many Galactosemia websites for extra support and updates. It really helps knowing that there are other families going through the same challenges as we. We are really looking forward to the upcoming Galactosemia Conference to learn as much about Galactosemia as possible and to meet other families.

Submitted by:

Terry

Virginia

Evan's Story

Evan was born in February of 1998 at Waukesha Memorial Hospital in Waukesha Wisconsin. He was our second child and was delivered on a Friday night. We had no idea that either mom or dad was a carrier for galactosemia. Our first child, Sara, was born without any complications in October of 1995. Shortly after Evan was born, I was sure I would breast feed him. Evan was fussy and did not want to eat. It seemed that something wasn't quite right with him, but I couldn't put my finger on it. He was 7lb 12 oz and 19.5 inches without any outward abnormalities. Newborn screening was performed as usual and he was sent home on day 2 from the hospital with only some newborn jaundice.

Evan continued to feed poorly and became more jaundiced. We brought Evan back in for a bilirubin level 1 week later and it was very high. Hospitalization was necessary in the NICU at Waukesha Memorial Hospital for phototherapy and treatment to bring down his bilirubin level, which had climbed to almost 20. I continued to try and breast feed him in the NICU, but he was still not interested in feeding. I became very worried when his bilirubin level did not come down at all after 1 day. He was on triple phototherapy and had IV fluids running. His doctor was "stumped" 2 days of hospitalization and his level continued to rise. A trial of formula was started and I stopped nursing Evan per doctor's orders, thinking it could be breast milk jaundice. Evan was released as his levels started on a downward trend. We still heard nothing from the State at this point about his newborn screening - we are going on day 8-9.

I continued to feed Evan breast milk at home since his doctor said it would be O.K. But, Evan was still not feeding well and continued to be fussy at the breast and irritable. Evan then began vomiting again and was brought back in to see his pediatrician. He was exhibiting signs of liver failure. Our doctor decided to call the State of Wisconsin Newborn Screening in Madison to see if indeed there were any abnormalities noted on the metabolic screen. After the call, the doctor informed us that Evan had indeed tested positive for Galactosemia. I have never been more frightened in my life. This was a big shock to us all!!! We still do not know why the State didn't notify us of Evan's galactosemia test!! It is possible that his blood test did not get sent out in a timely manner.

Of course, newborn screening was sent out again to confirm the diagnosis. We were thankful we found out what was wrong and immediately our son was placed on a soy formula - he was 10 days old. Within 1-2 days of eating soy, Evan began to improve significantly and totally recovered from the jaundice. Evan continued to grow normally and to feed well tolerating the soymilk.

This is how we found out about Evans galactosemia. We thank God everyday that he is here with us. We thought about pursuing the state and hospital, but never did. I am a registered nurse in Labor and Delivery and since this happened, I teach every one of my patients to follow-

up with their pediatricians about their newborn screening results. I stress the importance of following your gut feelings when something is not right with your baby. I have told our story to many people in hopes that they will tell others about the importance of early diagnosis and how critical it is to neonates born with galactosemia.

Submitted by:

Sandy

Wisconsin

Alena's Story

Our daughter, Alena, was born on November 22, 2003 in Portland, Oregon. In the two days we spent in the hospital after birth, she didn't eat much and would spit up constantly. The nurses decided to flush out Alena's stomach when she didn't stop spitting up. Despite the fact that Alena wasn't eating much, our doctor discharged us from the hospital.

At home, Alena was unable to keep anything down and had begun dry heaving. When she was 6 days old, we took her to see the pediatrician. Thankfully, our pediatrician had a "gut feeling" that something was not right and referred us to the hospital. Nobody knew what was wrong with Alena until that afternoon (the day after Thanksgiving). The result of Alena's newborn screening was not yet available. The hospital performed a spinal tap and started her on three different antibiotics.

The pediatrician in charge of the pediatric floor explained to me that a positive for Galactosemia was the same as hitting the jackpot in the lottery!! He hardly knew anything about Galactosemia, but told me that children with Galactosemia are prone to develop E. Coli Meningitis.

The results of Alena's spinal tap were received the next morning and Alena had E. Coli Meningitis as well as sepsis. On Saturday the infectious disease physician in charge told us how serious the situation was. Alena had a seizure and was transferred to the pediatric intensive care unit. These were the hardest days. Alena was only one week old.

Alena stayed for 3 days in the Pediatric Intensive Care Unit and then was moved to the Neonatal Intensive care unit. She was in the hospital for 14 days.

Alena is now 2 $\frac{1}{2}$ years old and is doing great. We took Alena on her first trip to Germany last fall and she very much enjoyed it.

We recently had another baby, Mia Rose Elizabeth, who was born on February 15, 2006. Mia Rose tested positive for Classic Galactosemia also. Both Alena and Mia Rose are doing fine. Ron and I are very proud parents.

Submitted by:

Beate

Oregon

Kyleigh Elizabeth's Story

October 21, 2004, we were blessed with a beautiful little girl we named Kyleigh Elizabeth. She weighed 6 lbs, 13 oz. We had difficulty getting pregnant and when she finally arrived, we thought her the most beautiful little baby we had ever seen. We were advised that breast feeding can sometimes be very difficult in the beginning, but we decided to give it a try. Kyleigh was very fussy during feeding, but we were determined to keep trying.

Kyleigh's first visit to the pediatrician determined that she had lost weight, down to 5lb 7 oz. The pediatrician assured us it was normal for a newborn's weight to drop slightly. She also indicated that she wanted to do an additional blood test. She said she had received an abnormal reading from Kyleigh's newborn screening tests on some disorder known as Galactosemia. We had never heard of this and asked what it was. The doctor casually told us, "Don't worry, it's just a diet thing, we don't think she has it anyway, it's very rare". It would take 3 days to obtain the test results.

Two nights later, we received a message from the doctor's office to call immediately. We did, and spoke with a different doctor who instructed us to take Kyleigh to Children's Hospital in Boston immediately. The doctor said a team would be awaiting our arrival in the emergency room. My husband and I are both police officers and are trained to deal with medical and other emergencies. This was totally different, we were numb.

We arrived at the hospital late at night and Kyleigh was immediately brought into the NICU where she remained for 5 days; her final diagnosis - Classic Galactosemia. We talked to "alleged experts" on Galactosemia, social workers, nutritionists, medical students. This was the longest and hardest five days of our lives. Kyleigh was hooked up to monitors and fed by the staff, we felt so sad and helpless.

It is now ten months later, and Kyleigh has also been diagnosed with Plagiocephaly, Torticollis, and Scoliosis. We are in Children's Hospital quite frequently. Kyleigh wears a helmet and has physical therapy every week. She has been through so much. We know how strong she is and that she is a fighter. She is a happy baby and laughs often.

So far, our experience has taught us that we are going to be the ones who are the experts on this disorder. We've experienced lab tests conducted improperly and a nutritionist who instructed us to feed her something with an extremely high galactose level. Researching this disorder and writing to PGC we feel we are not alone anymore, especially when we received contact from the President, Michelle Fowler, who has been very helpful and supportive.

We know we all come from different walks of life; however, we all have one thing in common, finding a cure or treatment for Galactosemia.

Submitted by: Kristine, Massachusetts

Adam's Story

Twenty-two years ago, as my obstetrician rolled the ultrasound wand over my gel-covered abdomen, I watched his face for a sign, suddenly his eyebrows squinched together and I knew my feelings were confirmed - I was having twins! When I was pregnant with my first child, I remember thinking, maybe its twins, but this time it was different.

As a writer and researcher, the first thing I did was to read everything I could about "having twins". Of course, this was before the internet. I diligently followed a high-protein diet. Not being a milk drinker, I forced down a glass at least once a day along with eating protein at every meal.

Allison and Adam were born on a Wednesday; each baby weighed more than 7.5 pounds; Adam's weight probably helped to save his life. I couldn't shake a feeling that something was wrong. Allison was born first, Adam was next. His Apgar score was low, but that's to be expected with a second born twin, the doctor explained. I wasn't so sure. The first time I held Allison, she squirmed and wiggled and gobbled down her formula. The first time I held Adam, he barely moved, he was so floppy that he had to be swaddled in his blanket. He was a face and was not at all interested in the bottle I tried to feed him.

I started probing the pediatrician and the nurses. I told them Adam didn't like his formula and they said "he'll get used to it". I actually asked the pediatrician to change his formula to soy (my daughter had colic and was placed on soy) and he refused.

When Adam became jaundiced, they put him under the bilirubin lights. I actually felt better; I had friends whose babies had jaundice. But, Adam continued to get worse. I kept asking why is he so listless, why can't he open his eye, why does he seem so tired. I was told that sometimes the bilirubin lights can have that effect on the babies. I asked if it was normal then why are the whites of his eyes so yellow?

When Adam was 4 days old, the pediatrician made an unexpected visit to my room during visiting hours. The minute I saw his face, I knew - my feelings were once again confirmed. He told us that Adam was very sick and there was something wrong with his liver and they didn't know what it was. Adam had been moved to an isolation nursery in case it was a virus, away from his twin, and the next day he was transferred to the NICU at Westchester County Medical Center, which was about $\frac{1}{2}$ an hour from where we lived.

The next day was Mother's Day. They brought Adam to me in his transport incubator. There were so many wires and tubes attached to him that all I could do was hold his tiny little hand in mine. The feelings that any parent experiences during a time like this are overwhelming, it felt like someone had just shattered our world. This didn't happen to people like us, this happened to other people. Our nightmare had begun.

The next morning, my doctor reluctantly released me from the hospital. My husband and I left with only one baby, who we dropped off at home with her older sister (2 year old Melissa) and their grandparents. We went straight to the medical center.

Life became a blur of days and nights, beeps, buzzers, tiny babies in incubators, other parents dazed as we were overworked nurses and doctors, and still no answers. They were all doing types of tests including a spinal tap. Adam wasn't taking his formula, they started him on antibiotics. I asked if Adam was in a coma, the doctor said "No, but he is very, very sick. There are babies here who are much sicker than your son and some that are not as sick, but your son poses the most interesting case to us". My mind locked with my husband's "an interesting case" - we squelched the growing rage at this man who was talking about our son.

On Adam's 8th day of life, they found the answer. An ophthalmologist who routinely checked the preemies eyes had checked Adam's and diagnosed him as having "Galactosemic cataracts". What we'd learned in college biology came back to us as the doctors explained this rare, recessive gene disorder. He was immediately taken off of his formula and started on soy.

Unfortunately, the doctor explained, we can't confirm this immediately. Why? Because they could not figure out how to transport Adam's blood to NYU, the only hospital in the area that did the genetic testing for Galactosemia. My husband said, give me the blood, I'll take it there. Oh no, that was against hospital policy. He then told them what they could do with hospital policy and we took the blood to NYU and confirmed the diagnosis the next day.

From that day on, I have relied on my feelings when it comes to Adam and my two daughters. I knew something was wrong from the beginning. There was nothing I could do about it then, but I never let a doctor, nurse, teacher, etc. ever discount my feelings again.

When Adam was 8 months old and didn't respond to his name, I took him to an early intervention program where he received speech, cognitive, and occupational therapies until he was 5 years old. At 7 years old, his special education teacher told me he would never read. I hired a private tutor (who works with him until this day) and he reads on an 8th grade level. I was told he would never swim - he did as I slowly let air out from his swimmies; he wanted to keep up with his sisters. I have not and will never take "no" as an answer; when it comes to Adam, he is one of the happiest people that I know and takes everything including his severe hand tremors in stride.

I have learned to listen to my heart when it comes to Adam, he has taught me and our family and friends so much. My advice to newly diagnosed mothers is "go with your feelings, you're probably right".

Submitted by:

Linda

Florida

Jack's Story

My name is Samantha. My younger son, Jack (5/31/05) has Classic Galactosemia.

Jack was born on a Tuesday in the early afternoon. He was a beautiful baby. He had a thick head of black hair, perfect pink skin, and a wonderful personality even from so early on. We stayed in the hospital for two days. The pediatrician thought Jack looked a little yellow and tested his bilirubin level, which was 6.6, but sent him home anyway on June 2nd. We were at home for 4 days when we went back to the pediatrician's office for a check-up. Again, the pediatrician was concerned about his color. I saw that he was yellow too, but, was he more yellow than when we left the hospital, who knew?

We were sent immediately to the hospital for a blood test. I dropped off my husband and my older son at the park and my mom met us at the hospital. When Jack's test results came back, he had a bilirubin level of 26.5!!! He was immediately admitted to the Neonatal Intensive Care Unit and there he stayed under the trip-lights with my husband or me standing a 24-hour vigil for the next two days.

Wednesday afternoon, June 8th, the neonatologist let it "slip" that he was concerned about how lethargic the baby was and the results of his liver function tests. Until this point, I had convinced myself that this was because his liver was immature, after all, he was born 10 days early. Besides, this was my baby, there couldn't be anything wrong with him. Nothing bad could ever happen to my children. But, after speaking with the doctor I began to have a very uneasy feeling. Almost 24 hours to the minute after speaking with the doctor, one of the nurses handed me a packet on the New York state newborn screening test. She told me maybe my son had one of the diseases that were being tested and if he did the results would be back soon. I felt dizzy, I thought I was going to pass out, but I knew I had to stay strong for my baby. As I was pulling myself together the phone rang, "Hello, yes, we have a Jack McHale, yes, hold on, and let me get you the doctor".

On the phone was someone from the metabolic treatment center from our county. The diagnosis was Classic Galactosemia detected by the newborn screening heel test. Jack was immediately switched to Isomil and we saw the difference in him within a couple of hours after that. Our NICU nightmare ended 5 days later when Jack was finally sent home.

The last year with Jack has brought us an endless stream of doctors, therapists, countless blood tests, endless label reading, and a lifetime of joy. My parents and I know more about Galactosemia and the diet than most nutritionists. Friends and family members have been more supportive than I could have hoped for. Jack's older brother, Sean, tells everyone he sees that Jack can't have certain things because they make his "bro-er" sick. Our babysitter has been amazing... she calls me every Thursday from the grocery store with lists of ingredients, squealing like a happy baby every time she finds something new that Jack can have. Jack is growing into the wonderful little person that he is. He is slightly delayed in OT, PT, and speech, but has been in early intervention since January 2006 and is doing beautifully.

Submitted by:
Samantha
New York

Tiffany & Andrew's Story

Tiffany was born on April 22, 1980. She was 7 days old when she was diagnosed with Galactosemia. It was touch and go initially. She was hospitalized for 14 days.

This was the start of the journey for us. We had never heard of Galactosemia and we knew nothing about it. This was the beginning of our learning process.

Andrew was born on June 28, 1986. He was diagnosed right away and this time we were more prepared.

Tiffany is now 26 years old; she graduated from a vocational high school program and works at a local supermarket.

Andrew is 20 years old and he is a sophomore in college and is looking towards a career in business.

Both Tiffany and Andrew have adjusted incredibly to their diet restrictions and have become admirable adults

Isabella's Story

Isabella was born on March 21st, 2005. She was born as a repeat c-section. I was glad to see immediately after her birth that she had all body parts present and looked healthy. I was reassured after initial and subsequent checkups by a pediatrician that she was doing just fine. However, my mother's intuition knew that something just wasn't right.

This little girl was almost always fussy and couldn't settle down and get a good night's sleep. She was keeping all of the other babies in the nursery awake. When she nursed, she would pull away and cry although she seemed starved. When I would try and lay her down she would start crying.

During the four days she was in the hospital she lost a pound and a half. The pediatrician kept assuring me that she was o.k. After returning home and on the 8th day of delivery, my mom took my other children out for an Easter egg hunt. I was thrilled as I thought this would be my first chance for some real sleep. About a half hour after falling asleep, I received a call from the pediatrician's office. He told me the newborn screening results were back and Isabella was positive for Galactosemia. He told me he was going to set up an appointment with a metabolic specialist and nutritionist.

I was still in a drowsy fog and could barely believe what I was hearing. Was I dreaming this? The doctor told me to stop nursing her and to try a soy formula. My other 4 children all nursed, so I didn't know the first thing about bottle feeding. Panic consumed me at the thought of Isabella's condition and her not getting adequate nutrition. I felt helpless.

There wasn't any formula in my town, so I called my sister-in-law and asked if she would go to a neighboring town and pick up some soy formula. I was overwhelmed with grief and apprehension. As it turned out, Isabella was in the pediatrician's office the next day because she was having bloody stools from the soy formula and was switched to Nutramigen.

Submitted by:
Kodi
Nebraska

Parker Wayne's Story

With a loud cry and shriek at 10:05 am, December 3rd, 2004, our son Parker Wayne was born. Parker was 3 weeks early and delivered via c-section due to mom's high blood pressure. He came out a mere 4lb 10oz, but seemed perfect to us. After a little cuddling with mom and dad, Parker was sent to the NICU where he would spend the next 16 days.

At first Parker seemed fine, small, but doing o.k. requiring no oxygen or IVs just an incubator to keep him warm. As the days passed, Parker became sicker and sicker. First he couldn't maintain his body temperature and then it was jaundice. On his 5th day of life the doctors thought he had caught a staph infection and did a spinal tap. Parker seemed to be getting weaker and weaker and even seemed to turn a grayish color. None of the doctors seemed to know what was wrong with him and everyone seemed concerned. As first time parents we were very scared and overwhelmed by the love we felt for our son. Yes, we saw the problems he was having and of course we were extremely concerned, but we were just so happy to have our baby here. We must have taken at least 20 pictures of him in the incubator. All of the nurses loved him and he quickly became the star of the NICU (beating out a pair of noisy premature twins and a little girl with a heart murmur).

On Parker's 9th day of life, the head of the NICU, Dr. Damor, decided that he might have something metabolic wrong with him and changed him to a soy formula. Overnight, we saw the difference!!! I remember coming into the NICU the next day and being shocked at how much better Parker looked. An hour or so later, Dr. Damor who had become very attached to Parker, came in smiling and in a French accent shouted, "Galactosemia". We were thrilled. No, Galactosemia is certainly something you shouldn't take lightly, but to us not knowing what was wrong was worse. Many people are sad on the day they find out their child has Galactosemia; to us it was a blessing because we knew he was going to be fine.

Submitted by:
Valerie
New York

Nicholas's Story

Nicholas was my first child and I was very scared about having such a huge responsibility. He was born by emergent c-section January 2, 1997. He was 9lb and 21 inches long. He looked perfect to me and to his daddy. Nicholas earned a nickname before leaving the nursery. He was trying to move on his knees it seemed and my dad dubbed him "scooter". The name still sticks today.

I chose not to breastfeed Nicholas (which turned out to be a blessing). He was placed on a regular milk-based formula. From the beginning, he wouldn't take his bottle and when forced to he would projectile vomit. I at first thought this was due to my inexperience as a mom, but even so I was very concerned; so concerned, in fact that I questioned our pediatrician, Dr. McCrary, pretty intensely during Nick's first 24 hours. The following day Dr. McCrary, not wanting to tell me everything would be alright, changed Nicholas from milk to soy. The nursery nurses were appalled. They told me it was ridiculous to change formula so soon. Anyway, Nicholas got better. He ate better, slept better, and quit vomiting. We were discharged home on day 3.

At home, Nick was a fussy baby and didn't like to sleep much, but he wasn't sick. On day 8 of his life, I received a call from the health department and was informed that Nick's newborn screening tests were elevated and showed a suspicion of galactosemia. They asked me to take him immediately to have more blood drawn. I had no idea what galactosemia was and found an old nursing book that had one paragraph about Galactosemia in it; after reading that I began to cry.

I took Nicholas and we drew blood on him. Dr. McCrary called me and we went to see him. He explained to me what galactosemia was. He had only had one other patient with the disease, but researched it and gave me some information.

Later, we were called and had to go see a metabolic geneticist in Jackson, MS. There they tested myself and my husband and drew more blood on Nicholas. They tested Nicholas' eyes. They gave us a list of food that Nicholas could eat and what he couldn't. What he couldn't eat was longer than the list to eat. I began to wonder how I was going to keep him alive. The geneticist, Dr. Bock, also gave me information about "Parents of Galactosemic Children".

I contacted someone from the organization and it was a pretty gruesome conversation. I didn't get any words of encouragement or hope, just the complications of seizure, mental retardation, learning disabilities, etc. This was my lowest point. I was at home alone when I made the call and became very emotional. I called my husband (at that time we owned a restaurant) and he came home.

Our pediatrician, Dr. McCrary happened to be eating at the restaurant that night and upon hearing that Mike came home to console me, gave me a call at home. He was a godsend. He was able to bring me back to reason. He also had the woman who's child he had treated with galactosemia call me the next day.

After that, I called or contacted anyone I could for information. One day I had the great good fortune to e-mail Susan Dudley, Christopher Mannix's mother. She wrote me the most wonderful, uplifting, compassionate story of her son that I began to feel hope for the first time. She wrote "I wish I could have seen the beautiful, intelligent, seeing, little boy my son would grow into" (I'm quoting this from memory).

I didn't know at that time that her son had lost sight in one of his eyes. She had the "good sense" and compassion to only share with me what really mattered. Those words were music to my ears and I will always be grateful to her for that. I quit reading research on the subject (since all of it's bad and it made my stomach hurt) and just started loving my son.

I attended a conference in Austin and finally got some real information about galactose content in foods and substitutions, etc.

Today Nicholas is 9 years old and perfectly normal in every way. He hasn't exhibited any signs of learning disabilities or delays of any kind. He's not the best student, but also not the worst. He is actually reading above grade level. God blessed us, through Dr. McCrary, because Nicholas never got sick. He had a really high Gal-1-P level upon birth (56), but only received 6 oz of milk his whole life. Ironically, if I had attempted to breast feed him, Dr. McCrary said he would have sent me home and said keep on trying. God works in mysterious ways his wonders to perform.

Submitted by:
Kelley
Mississippi

Lexi's Story

Lexi was born by planned c-section on September 25, 1998. Because I knew that we had a 25% chance of Lexi having Galactosemia I restricted all milk from my diet while pregnant with her. When she was delivered, Teresa, a metabolic nutritionalist came down from Jackson to obtain a blood sample from Lexi so we could test her enzyme activity right away. Lexi was put on soy milk from birth.

Sure enough, Lexi also has Classic galatosemia. Even though I restricted milk from my diet her level was still 13 and she was slightly jaundiced. We stayed in the hospital 2-3 days longer than usual because of hurricane George that visited us. We went home to candlelight and no air-conditioning.

When Lexi was 30 days old, she contracted viral meningitis. It was not e-coli. She was hospitalized and treated for 5 days. She recovered, but had a critically low H&H while in the hospital (7/22). We were unable to find anything wrong with her to explain her anemia and it eventually improved. Lexi came down with RSV at 90 days old and was hospitalized again. She has had multiple ear infections and received her first set of tubes at 5 months old. Lexi seems prone to infections (esp. viral) and we tested her immunoglobulin levels, but they were normal. Because of her ear infections, Lexi's speech was unintelligible some of the time. She underwent speech therapy when she was 3 years old and her speech has improved dramatically since then.

Lexi had a little trouble in the 1st grade and was put in Title 1. This is a federally funded program that gives an additional 30 minutes of reading, spelling, and phonic instruction daily. She did very well and her teacher doesn't think she will need the extra help next year. She seems to have a little trouble focusing, which I think has a lot to do with her outgoing, exuberant personality. Overall, she is a normal, beautiful child. She likes to eat however, and gets a little frustrated when she can't eat what her friends are having, but we're working on that attitude.

PGC has been very helpful to me with this disease, since most doctors and everyone else know nothing about it. I am very thankful to have been so blessed with two beautiful Galactosemic children.

Submitted by:
Kelley
Mississippi

Sarah's Story

Sarah was born on May 21, 1999 in Grapevine, Texas. She was our first child. The difficulties we experienced the first week of her life were assumed to be a learning curve. I was nursing Sarah, but she kept throwing up after she would eat. She was very fussy because she was hungry. The doctors thought she had reflux and had me put cereal in her bottle. I even went to a lactation consultant to make sure I was nursing correctly. Everything seemed o.k.

One week after Sarah was born, I received a phone call from her doctor. He said the results of her newborn screening showed she may have galactosemia. I was told to put her on soy formula immediately and bring her back to the doctor for a re-test.

I remember that day like it was yesterday. My family was in town for Memorial Day and they were very supportive. I was very confused and worried. I didn't understand what galactosemia was or why Sarah had it. I couldn't even pronounce the word.

After we got Sarah on soy formula, she was a new child. She was eating, sleeping, and growing. She became a very happy baby.

Sarah has continued to be a wonderful child. She is beautiful and loving. She loves babies, animals, and swimming. She also loves to read and go to school.

Sarah has a younger brother, Ethan, who is 5. We struggled with the decision to have another child. God took the worries into his own hands and I got pregnant anyway. We were careful to give Ethan soy formula until the results of his blood tests were back. I remember driving home from the hospital and getting the phone call that Ethan did not have Galactosemia. We were very relieved.

Sarah, Ethan, Mark and I are moving to Decatur, TX where we will surround Sarah with cows, horses, goats, dogs, and cats. She wants to be a veterinarian when she grows up. We'll see how she does on our farm.

Submitted by:
Jo Beth
Texas

Brian's Story

Bryan was born on June 23rd, 1995 and weighed 7lbs 12oz. The pregnancy was normal except that he was 11 days late. The delivery was normal, but his umbilical cord was wrapped around his neck 4 times. All that aside, we were the proud first-time parents of a beautiful baby boy.

After a few days (while still in the hospital), Bryan developed jaundice which is not uncommon in newborns. He wore a light belt for a few days to treat the jaundice. When Bryan was 7 days old, the jaundice seemed to be diminishing and we were released from the hospital.

The next few days we were settling in as a new family. However, something didn't seem quite right with Bryan. I had been nursing him from the earliest time that I could. He did some spitting up, but as a first time mom, this seemed normal. After we were home for 3 days, we took Bryan back to our local hospital for a check-up. He seemed more jaundiced and lethargic. His spitting up and vomiting was more frequent and projectile.

At the hospital, the nurse weighed him and noted he was down to 6lb 5oz. She sent us to the emergency department to have some blood work done. She felt he looked quite jaundiced. The lab ran his tests twice because the technician had never seen a bilirubin that high. Once the doctor had the results, Bryan was immediately transferred by ambulance to Sick Kids Hospital in Toronto. Once there, he was hooked up to machines and monitors in the NICU. It was quite disturbing to see our little baby among the many wires. He was very close to needing a blood transfusion. I was told to pump and store my breast milk until they could finish their assessment.

Within 24 hours of our arrival at Sick Kids, we were informed by the doctor that Bryan had galactosemia. It was a lot to process, Galactosemia was an uncommon, metabolic, genetic disorder, and the most common treatment was a restricted diet. He was not allowed breast milk only soy formula. Over the next 4 days they monitored Bryan and when he was 2 weeks old they released us from Sick Kids.

We have been fortunate to not repeat the ordeal of Bryan's first 2 weeks of life. Things have only gotten better. Bryan is being followed by Dr. Annette Feigenbaum at Sick Kids every year. Our appointments are always positive. She is extremely pleased with his progress; at our appointment last November, she referred to him as "our little miracle". Bryan has just turned 11. He does struggle with some academics, but his strength is in athletics. His passion is hockey and he has played it since he was 4 years old.

He received "Best Defenseman" for his Atom hockey team this past season and will play peewee minor hockey this fall. He is in his 3rd season of baseball and received MVP and Most Sportsmanlike awards in past seasons. We are very proud of his accomplishments.

Bryan is a very caring and sensitive boy who does his best to please everyone. He is very energetic. People are amazed when they realize that he has a genetic disorder. We are very fortunate and thankful that we have been blessed with him. Looking back, those first few weeks were very fragile, but we are well aware that Bryan's galactosemia and good progress are blessings in disguise.

Submitted by:

Sue

Canada

Sarah's Story

In 1975 we gave birth to our first child, Sarah. The pregnancy was uncomplicated and the delivery the most exciting event of our married life. She was perfect and I was determined to do everything exactly right. She went to the breast eagerly. We had one of those "progressive" family doctors who insisted on seeing his newborns back in 1 week (as opposed to 6 weeks). He knew something was amiss immediately - this beautiful baby was jaundiced and wasted looking. Her bilirubin level was alarmingly high and after a call to Children's Hospital in St. Paul, we made the anxious 50 mile drive north.

I was grateful to give over my sick baby - she lay so quietly in my arms, eyes closed. I felt the assurance that she was in the right hands and on her way to good health. Later that night, we had the diagnoses; galactosemia. At that time, there was no testing at birth for galactosemia so we were lucky to have the diagnosis made so quickly. Sarah spent 3 weeks in the hospital and finally arrived home at her birth weight - which seemed chubby at 6 lb, 10 oz!

It took another 3 months for her liver function to normalize; she had cataracts; otherwise she was thriving on her soy formula and we had begun the lifelong learning that accompanies living with a child who has a chronic condition. We have certainly had our ups and downs over the years. Sarah has a learning disability, but graduated from Business School. She is still determined to graduate from college. She works in customer service for Midwest Wireless in Mankato and owns her own home. She has been responsible for her diet since an early age is loving living in the 21st century with the advancement of soy products.

Pearls of wisdom from our 30+ years with galactosemia:

1. We were active participants in the development of the neonatal test for galactosemia. We offered Sarah as a test subject when she was a year old.
2. Prenatal testing has certainly come a long way, when we had our 2nd child Paul, both dad Jim and Sarah had to have skin biopsies to confirm the carrier and affected status to compare with the amniotic fluid; a painful way to make a diagnosis. Paul is neither affected or a carrier, a perfect genetic family.
3. Being proactive for your child is often a tough, uphill battle.
4. Sometimes your child doesn't want to be "special".
5. It definitely gets old explaining that galactosemia is not a milk allergy, but it's easier to explain it that way rather than watch peoples eyes glaze over.

That day in June, 1975 changed our lives forever - it will always remain the most wonderful day of our lives.

Submitted by: Mary Jane, Minnesota

Jacob's Story

Our first child, Jacob, stopped eating, turned yellow and became very lethargic at 3 days old. We couldn't get in to see a doctor, but the nurse told us over the phone that jaundice was normal in newborns and to put him by the window. When the situation grew worse, we took him to the emergency department. They put an IV in Jacob's scalp and tried to stabilize him before being transported to a NICU 2 hours south at Landstuhl Air Base, Germany. It was there we learned that Jacob would need a blood transfusion. They didn't hold out much hope for him even making it through the night. We were far from our families and friends and this was a huge blow to us, physically and emotionally. Gradually, however, Jacob's condition grew more hopeful. In the meantime, more tests were performed and finally a diagnosis of classic galactosemia was obtained. Because his bilirubin levels had been 44 there was brain damage done to motor areas of his brain, thus he was diagnosed with cerebral palsy. He is now almost 9 years old and continues to surprise us each year with how well he overcomes his physical circumstances. We have learned so much from him about determination and not giving up.

We were blessed with a little girl, 3 years after Jacob. Of course we were now armed with knowledge and information, so we could make the right decisions about her neonatal care. She was started on soy formula from the beginning as a precaution and her test results also came back positive for galactosemia. She is a normal developing 5 year old and is getting to be really helpful to me taking care of Jacob and learning about their diet. She does have a mild speech delay for which she receives services in school.

Submitted by:
Natalie
Alabama

Landon & Paden's Story

Our galactosemia story of horror is a little different than most. Our son Paden, who is 2 years old, never spent a day in the hospital (except for the 2 days he was born), or a day in NICU. You see he owes his life to his older brother Landon. Landon was our first born son whom had dwarfism, a hole in his heart, and an enlarged right ventricle, NEC, galactosemia, along with a lot of other problems. Landon died when he was 15 days old and the doctors all differ on the cause of death. We have come to the realization that Landon was not meant to be with us on this earth for longer than those 15 days and we would like to believe that it was all for his little brother.

When Paden was born he was immediately started on soy formula. The newborn screen was done and came back positive and he never missed a meal. Paden is doing very well now. His motor skills are great, his speech is lacking in excellence, but is average for a 2 year old and he seems to be acceptable of his diet. We like to believe that he is doing so well because his brother was sent as a guardian angel to warn us about galactosemia ahead of time.

Submitted by:

Kellie

Texas

Alannah & Isabella's story

Our first pregnancy was difficult. Jacqui was overwhelmed with sickness and migraines nearly the entire time. Our twins first revealed their impatience by insisting to come into our world early. Alannah and Isabella came by emergency c-section on February 14, 2001 at the end of the 25th week of gestation. Alannah weighed 1lb 6oz and Isabella weighed 1lb 14oz. They were sent to the NICU at Virginia Baptist Hospital where they stayed for the next few months.

The initial result of the newborn blood screen test came back positive for Galactosemia. I still remember the doctor vividly as he explained that the screening result was likely a false positive and in his greater than 20 years in practice he had never seen a child with Galactosemia. Luckily, at this time, our girls were still getting their nutrition intravenously. A follow up blood test was performed to verify the results. About a week after the 2nd test came back negative, much to our relief, but a problem was noted with the results. It seems that prior to the blood test, both girls were given a blood transfusion, which placed a big question mark in our minds concerning the validity of the blood test results. At this time it was assumed to be negative and the children were started on breast milk. A geneticist was consulted a couple days later and the girls were quickly taken off of milk and started on soy based formula. This was all done as a precaution because the staff didn't really think the children had Galactosemia.

We needed at least 30 days with no blood transfusion to get accurate results from the blood test. Unfortunately, each girl had several transfusions during their stay in the NICU. Jacqui had been saving breast milk during the kids stay with hopes that could eventually have it. Isabella came home a little over a week before her sister. We still had not gotten an accurate blood test, but the staff felt it was OK to start feeding them milk. Isabella began to nurse prior to leaving the hospital and Alannah was given the saved milk. After 1 week, one of the neonatologists noticed Alannah was not gaining the weight as she should have. Since Alannah's blood could not be tested, due to recent blood transfusions, a urine test was performed. The results of the urine test showed abnormal amounts of proteins indicating a possible metabolic problem. Armed with these results, the girls were put back on soy formula until a dependable blood sample could be obtained. After both girls came home they were taken to a genetics clinic at the University of Virginia. Blood was taken and in August, 6 months after they were born, Alannah and Isabella were officially diagnosed with Classic Galactosemia.

The twins are now 5 years old. A result of coming into our world so early is that they both have cerebral palsy. They are progressing well in school and outpatient therapy. Their younger sister, Amelia, does not have Galactosemia, but may be a carrier of the gene. God has richly blessed our lives with our children. We give all our thanks to him.

Submitted by:

Jacqui

Virginia

Living with Galactosemia

Sean- New Jersey

My name is Sean and I am four years old.

What does galactosemia mean to you?

I am not allowed to have dairy. No butter, cheese, some breads, milk, chocolate milk or ice cream. I would get sick if I had dairy. I have to bring my own snacks when I go places.

What do you like to eat?

Rice milk and waffles that Mommy makes. I have special chocolate chips and special cheese and special pizza.

What are your favorite things to do?

Playing my video games and playing on the computer. I like to play outside and play soccer and I like swimming too.

Living with Galactosemia

Nicholas- Canada

I am not upset that I have galactosemia. I do not let it bother me at all. I just know I can't eat certain foods. My friends always tell me that they feel sorry for me because I can't eat foods with cheese or chocolate. But to me, I've never tasted them so I don't know what I'm missing. I admit sometimes it's hard to go to parties because I have to bring my own food in case there is nothing safe for me to eat. My friends are good to me because they always remember to bring safe candy if I can't have the treat they bring to school or a party.

I am 13 years old and know what I can have just by reading labels. I never eat anything if I don't know the ingredients. Sometimes it's difficult to eat at restaurants because I have to ask too many questions about the food. I can't always order what I want, but I don't mind. When I was younger, I was afraid I would get sick or die if I ate something with milk in it. My mom told me that would never happen so I wasn't scared anymore.

I have tremors that make my hands shake. This makes my hand writing a little messy, but I'm thankful for computers so I can type most things. I had an occupational therapist work with me at school to show me exercises to strengthen my hands to overcome some of the shaking. Now, I mostly do push ups every morning to strengthen my hands. I also play hockey, baseball and do karate which helps with the tremors. Music is my passion and I play the piano, guitar and drums. That is really good therapy for my hands. I do my best to not let it stand in my way. I actually forget that I have galactosemia and live my life normally each day.

I have to visit the genetic clinic at the Hospital for Sick Children in Toronto, Canada once a year. I have to have a blood test to check my levels and that is probably the worst part of having galactosemia since I don't like needles.

I don't think growing up with galactosemia is all that bad. There are so many new lactose free products that I can eat; such as margarine, ice cream, soya milk and other treats. I just have to be very careful and read a lot of small print labels.

My mom makes all of my birthday cakes and they are the best. My friends want my mom to make their cakes too. My dad and sister are also very supportive of my diet. My sister trades Halloween candy with me and will eat popsicles instead of ice cream in front of me. My dad teaches me to read labels in the grocery store and quizzes me on the safety of the ingredients.

My grandparents, uncles and aunts always make sure there is food I can eat when I visit their homes. I am lucky so many people care about me. I think my life is great. Growing up with galactosemia has been manageable for me and I hope other galactosemic children feel this way too.

Living with Galactosemia

Landon - Mississippi

I am Landon. I am 6 years old and I go to school. I will be in 1st grade in a couple of weeks. I have had Galactosemia for over 6 years.

I have 2 sisters; Tori and Tela.

I cannot have cheese, milk, dairy, whey, tomatoes, and a lot of other things. I am OK with the things I can not have. I do have some cheese and milk that is safe for me, but I don't eat the cheese that much.

I do not want anything that I cannot have because I am Galactosemic. If I ate something I was not supposed to have Mama said it would build up in my system. If that happens it would be really bad for me.

My friends always ask me about my medical bracelet. I tell them it is my medical alert bracelet about Galactosemia. I tell them all of the stuff I can not eat. They just do not say anything when I tell them.

Sometimes people offer me something to eat and I just tell them "no thank-you" because I cannot have that.

When we go out to eat, I cannot have some stuff on the menu. I can only have a little bit of stuff. I could have everything if they did not put all that stuff in food that I cannot have. Like when we go to the place that is hooked to the mall, I can only have 2 things. My sisters can have everything at every place. Some places will cook me some special stuff. Some places give me bread that doesn't have butter. Papa cooks me meatballs just for me and a bunch of them so Mama can freeze them for me. Then Maw Maw cooks me pancakes cause Mama don't know how to cook them. I think Mama freezes those too, but I am not sure, I just know there is a whole bunch of them and we have to let them cool a long time before we put them in the bag thing that I see Mama get out of our freezer when I want one. I always ask Mama & Daddy if I can have something.

My favorite food is noodles and my favorite drink is angel milk. Angel milk is my milk with strawberry quick in it. Mama told me it is angel milk because it has been kissed by an angel. I like my soy and rice milk because I have to drink so much everyday.

My favorite thing to do is play the computer when mama lets me. I play baseball, soccer, do gym and like to swim too.

I am going to always be OK with Galactosemia because I know I am special and there are other galactosemics people too. It is not just me.

Maybe one day it will be better because my Mama sure does spend a lot of time doing PGC stuff and talking to Ms. Angie!

Living with Galactosemia

Kyle- Colorado

There are sometimes when I like having Galactosemia and there are times when I hate having Galactosemia. The times that I am happy with having Galactosemia, are when I am thinking of how mentally strong I am because I don't sneak foods that I can't have. Also our diets make us eat really healthy food so we can live longer.

The times I hate having Galactosemia is when I am at a family or friend party and they eat cake that I can't have; it makes me very angry. I also hate Galactosemia because when I am reading the ingredients list and I get down to the last line of ingredients and it says milk or may contain milk. For me, my confidence increases for every line of ingredients that I read and I don't see any milk ingredients and then to have my confidence destroyed because it may contain milk, you know it makes me pretty mad. The thing that hurts the most is not being able to eat the same foods as your best friends.

In conclusion, Galactosemia has its ups and downs.

Living with Galactosemia

Sharon - California

As a child, I had no negative feelings about having Galactosemia. It was who I am. I was unable to eat dairy products. Big deal! My mom was a stay at home mom who prepared all of our meals. She had complete control over my diet. That was fine with me. She cooked and baked a variety of foods I loved. We rarely ate meals outside of the home. It was impossible to know with 100% certainty if foods from a restaurant did or did not contain dairy products.

I never felt bad for not being able to eat what others could. My parents taught me early that some things others could eat were poisonous for me. That was, and still is, a fact. But, there were other things I could eat. If a classmate or a member of my family was eating chocolate cake, for instance, I had angel food cake. I ate Jell-O instead of ice cream. There was always something for me to eat. Not just at my own home, but at the home of other family members.

Holidays and other family get-togethers were never a problem. I have an extended family that has always remembered to prepare something special for me. My grandmothers, aunts, cousins always set aside mashed potatoes, sweet potatoes, breads, and desserts without milk ingredients. It's the same today. My extended family is large and everyone brings a dish. If that item contains milk, butter, or other dairy products, a small portion without dairy is set aside for me.

As a child, I did visit doctors at Children's Hospital of Los Angeles, often. For me it was a treat. I knew I had the best doctors in the world and I loved them. When we visited Dr. Richard Koch, Dr. George Donnell, Dr. Karol Fishler and Elizabeth Wentz, the dietician, I honestly thought they went to work just to see me!

Things changed drastically when I was 19 years old. That was when I was told I'd never have children. It feels like yesterday, even though it's been 27 years ago. It's still almost too painful to think about and almost too difficult to write about.

I was preparing to leave home for college. Going to college was much more than a dream. It was part of my life's plan. As a young child, I made myself a very short list of things I HAD to accomplish; #1-attend college, #2 - live in Orange County (where I lived until age 7). I was two weeks ago from both of these goals; I was going to California State University in Orange Co.

Although plan number 1 & 2 were in reach, plan #3, being a mother, died in an instant. I received an unexpected telephone call from a doctor at Children's Hospital, who told me that the longer I waited to start a family, the less chance for me to get pregnant.

Her message hit me like a bolt of lightning. I remember giving the phone to my mother. I didn't want to hear anymore. I didn't have a boyfriend. I had never seriously dated anyone. Plan 3 was supposed to happen after college. The thought of not having a child was never a plan.

During some of those prior visits to Children's Hospital, Drs. Koch and Donnell would sit in a large conference room with my family and discuss the possibility of my children having Galactosemia. I never learned in that room, that I may never have a child.

I do not blame the doctors. They were as surprised as me to learn I might not be able to conceive. However, I was only 19 years old. I felt more pain and concern about my future than any doctor could imagine. To say I cried often and a lot would be an understatement. I felt as if my heart was breaking. I could do nothing about it.

As a Monday morning quarterback will do, I think of things in my life I could have changed. Sometimes I wonder if I should have postponed my move to college. In 1979, I went ahead with my scheduled move because I did not want plans 1 & 2 to die with plan 3, but my heart and mind wasn't fully in it. I graduated in 1983.

For me, infertility has been a painful challenge. I have always loved children and believed I would have been a wonderful mother. I knew I would have to marry a man who loved children, but the gnawing question still remains; "why me"? "Why doesn't God want me to have children"?

I believe that menopause and infertility had a disastrous affect on my self-esteem. I struggle with low self-esteem daily. I don't remember having low self-esteem as a child. I believe it developed in my late teens and early twenties when I went through menopause; until then, I thought I could do and be whatever I wanted in life.

Today I'm 46 years old and a substitute teacher; because of my age and my job, I longer want children. Yet the pain of infertility still exists within me. For me, it's a cruel element of Galactosemia.

I understand that some patients with Galactosemia are more severely affected than others, and I know that thanks to my mom, my doctors, my family, and God I am doing far better than most.

Living with Galactosemia

Adam- Florida

"Pass the Cheese"

My name is Adam. I am 22 y/o. I was born in Middletown, New York. I have a twin sister Allison and an older sister Melissa - I call them the "sisters".

My parents found out that I have galactosemia when I was 8 days old. My parents told me I was very sick, but I don't remember. I am very afraid of needles and I think it was because I was stuck a lot when I was born.

I don't like having galactosemia. I don't think anyone likes to have a disease. I can't have cheese or ice cream or anything dairy. When people ask me if I want pizza or ice cream or stuff that has dairy in it, I tell them I can't have it because I'm lactose intolerant because nobody knows what galactosemia is and it's too hard to explain. If people ask me what happens if I eat dairy, I tell them I will get sick.

I like it when my parents or sisters tell people about galactosemia because then I know people will understand me better and why I'm on a special diet.

People who don't have galactosemia really don't understand what it's like. If I could eat dairy, I would eat cheese, especially on pizza and Big Mac's and I'd get a cheeseburger too. It would be really nice to have cheese.

When I go to restaurants, the servers usually understand when I tell them I can't have dairy, sometimes they don't though and if my parents are there, they get annoyed with the server.

When my family or other people eat things at the table that I can't eat, it bothers me, but I never say anything, but I should especially if it's my family. My family always gives me something that's almost the same, but pizza with only sauce and pepperoni is not the same as pizza with cheese. When I was younger, I used to feel mad about it, but I didn't say anything; now, I only feel mad sometimes. If I get something really good like dairy-free chocolate it doesn't bother me that much.

One thing I need to learn is how to read labels, but I don't feel like learning it yet, because my parents do it for me.

If there was a cure for galactosemia, the first thing I would eat would be cheese, especially on pizza.

Living with Galactosemia

Christopher - Virginia

My name is Christopher and I have galactosemia. Galactosemia is a rare genetic disorder that I was born with. It means I cannot have any milk products. It is much like being lactose intolerant, but it's different because my enzymes can't break down lactose. I am missing the enzyme that breaks down galactose. Galactosemia is more serious than lactose intolerance because the galactose molecules get inside cells all across my body instead of just targeting one area of it. Because of this, I am blind in one eye.

When I was born on June 7, 1993, the doctors told my parents I was "perfect". When I was less than 2 weeks old, though, the doctors and my parents were worried. I was jaundiced and was not gaining my birth weight. I went into the hospital overnight and was given a blood transfusion. I seemed so much better my parents took me home again. I went to the doctors several more times because I was skinny and yellow and throwing up a lot. Finally, when I was 5 weeks old, the doctors figured out what was wrong. Dr. Liebowitz saved me! He told my mom to stop breast feeding and put me in the hospital for a week. That's when they discovered I was blind in both eyes. One eye cleared up because it was only a cataract. The other had a vitreous hemorrhage which has left me permanently blind.

I don't remember any of this, but I do remember going to a PGC conference in Ohio and one in Georgia. I loved Ohio because all the people who got to know me liked me. I was only 5 and I liked getting up in front of the microphone.

Without having any milk products - that includes cheese, ice cream, tomatoes, and beans - I have to check the ingredients on many store items. But, there's a lot of good stuff I can eat. I like burgers and fries, spaghetti with tuna instead of spaghetti sauce, and sorbet. I just finished 7th grade. My favorite subjects are science and composition. I like to be in plays and I take Tae Kwon Do.

The X-Men are mutants who get mutant super powers when they hit puberty. Since I am a mutant who just turned thirteen, I'm waiting for my powers to come. I think my power is the ability to make people laugh.

Living with Galactosemia

Holly- Massachusetts

My name is Holly and I am a 28 y/o with Classic Galactosemia. I was born in Largo, FL and now live in Boston, MA. I was very sick when I was born and almost died. My mom said I cried a lot, but really I don't remember.

Sometimes it's hard having a special diet; especially when I'm with my friends or going to a party. It takes a lot of planning and phone calls to make sure I can eat what is being served; in most cases I have to bring my own food. I think my friends feel sorry for me that I can't eat cheese pizza and junk food. If my friends ate more fruits and vegetables as I do then everyone would be skinny like me.

Eating out is fun, but not always easy. Once, my mom and I went to a restaurant and the owner asked us not to come back! My mom asked him lots of questions about the food. I guess that made him mad. I try to eat at restaurants where they know me and are familiar with my diet. It makes it easy for me to order what I like. I eat at Chili's a lot and order the fajitas, but I need to bring my own taco shells.

I don't mind telling people I have Galactosemia. There are other people in this world with conditions much worse than mine. Although some things are harder for me to do, I just concentrate and work hard to accomplish them. Every Saturday night I go to bingo with my friends. It takes me longer to find the number so I have to work hard to keep us with everyone and my friends help me out too.

I also have tremors that make it difficult for me to do things like writing, holding my coffee cup and cooking. I'm disappointed that I can't do arts and craft activities like painting and pottery. I have others help me out with the things I need and everything works out okay because I can do lots of other things. I can swim, bowl, play softball, and ski.

Overall, Galactosemia isn't all that bad. I know my weaknesses and learn to overcome them with patience and assistance from others. I know it takes me longer to do some things, but I've learned not to get frustrated. I focus on the things I can do and try not to think about the things I can't. That's how I've learned to live with Galactosemia.

Living with Galactosemia

Kimberly- Missouri

I was born before the doctors could do ultrasounds to find out if the baby was a boy or a girl. My parents were told that I was going to be a boy. They bought boy clothes and had to exchange them for girl ones. I am the 4th of 5 children so when I went home from the hospital my parents knew that I was not supposed to be throwing up that much. They brought me back to the hospital. I had jaundice. I had a high enough bilirubin level that I needed a blood transfusion. I lost weight and had failure to thrive. No one knew what was wrong with me. I was even put on different formulas. Since the doctors at the hospital where I was born in Garland, TX did not know what was wrong, I was sent by ambulance to children's hospital. When I was ten days old, someone brought a medical book to my parents. This person had been doing some research. They said they were 99.9% sure that I had Galactosemia; so, I was put on Isomil.

Growing up I had a lot of learning problems. It seems like I always got C's and D's in school. I took a test in 2nd grade to see if I should be in special school. I passed the test by one point so I never went to special school. My siblings seemed to do well in school. Some of them were in gifted and/or honors classes and they all got better grades than I did. In high school I took choir and childcare courses and I actually did better in those classes. But, after going to some of the PGC conferences I wonder why I never received any extra help. I know others have actually had tutors, etc.

My parents taught me what I should not eat, but the list was always changing. My parents also taught my siblings what I should not eat so that they would not feed any of it to me. My parents taught me how to read ingredient lists. Eventually, I was skimming those lists because I knew what to watch for.

When I was in school, my parents would have me bring my own sandwich and other lunch items in a brown paper bag. I never once had a hot lunch and I was a little tired of sandwiches by junior high. In High school we had extra lunch choices such as a salad bar, vending machines, and there were always French fries as a choice so I still brought my lunch most of the time, but sometimes I just wanted salad or French fries. Also in elementary school, my parents usually gave the teacher a big bag of Starburst candy at the beginning of the year. The teacher would give me some of the candy during birthday celebrations, other parties, or just when the class was having an extra snack.

My siblings would all tease me with food; I don't know who was worse. My brother would eat rocky road ice cream and tell me I could eat it. Then he would tell me to go to the road and start eating the rocks. I actually believed him the first time. My youngest sister maybe did not realize she was teasing me, but she was always so picky and it seemed like all she ever ate was chocolate, pizza, donuts, ice cream, cake and other foods that I could not eat. She was always eating it right in front of me (sometimes putting the food right in front of my face) and I kept thinking if that is all she eats then it must be good. But, they also wanted to protect me. I was shy and my siblings would tell other people "she can't eat that". At pot luck dinners or buffet restaurants they wanted to help me find foods that would be ok to eat. Now that I live away from them they have forgotten what foods are OK and they also remember how my lists used to change. If I go to stay with them at their houses they usually ask me things like brand names of safe bread or margarine because they want to be able to have safe foods for me.

Living with Galactosemia

Lexi

Hi, my name is Lexi and I am 7 years old. I have classic galactosemia. I hate galactosemia. All of my friends get to eat milk and I don't. When I go somewhere new, they bring something that I can't have.

Living with Galactosemia

Martial - Texas

My name is Martial. I live in Katy, Texas. At school i participate in band, play soccer and am also a proud owner of an XBOX 360. Some would think of me as a normal kid, just making my way through high school like all of the others out there, but little do they know I have a medical disorder - Galactosemia.

Everywhere I go, whether to a friend's house, the movies, a band field trip or just down the street, I have to go through the trouble of reading ingredients labels of every single edible object that enters my mouth. Not exactly fun. If I go to a party and there is pizza, ice cream, cake, you name it, I can't eat it. I get to sit there for 20 minutes and hear "this cake is so good". So I have to bring my own food pretty much anywhere I go, most especially if it's an over-night thing.

I personally hate Galactosemia. It's my biggest enemy. I have to remember about 30 things that can't be in the foods I eat, and that's the best part. It's much harder going up to a complete stranger and asking about their food. For example, it could be at a restaurant, fast food or a sit down type, I have to question the waiter on every single ingredient in my food. Sometimes it comes later than everyone else's and they have to wait on me. What fun.

It's not fun, but it's the way it is and there's nothing I can do about it; for now at least. So I have to live with it and deal with it. I have no choice. But, don't get discouraged too bad if you have it. Since we were born this way, let's make the best of it. If you think about it, there are a few ways around it. "No cake" is a practiced rule, yes, but my mom can make "safe" cakes, cookies, and pancakes using milk substitutes, as many Galactosemic families do. So if you think about it, I guess we aren't missing out on too much as far as what we can actually eat.

The reason that living with Galactosemia is hard isn't because I'm "missing out" on all of the tasty foods that non-Galactosemic kids can eat, because really I'm not. It's having to go places and eating out of town and such. I'm not saying I hate leaving the house, heck, we're leaving for Florida in two days and I can't wait. I think that going out of town's worth dealing with my diet. Especially band trips...and on that subject, I got 1st chair in the top band this coming year. I'm in 10th grade and I beat out all of the 9th, 10th, 11th, and even 12th graders for my 1st chair position. I'm not saying this to brag, but to give hop to all of the Galactosemic families out there. When I was a baby, the doctor was very negative telling my parents about Galactosemia. But, doctors don't always know what they're talking about. I'm doing just fine. So don't lose hope. No matter what the doctor says.

So yes, living with Galactosemia is a pain and I hate it, but that's the way it is, so I might as well live the best that I can. I encourage you all to do the same.

Living with Galactosemia

Nicholas- Ocean Springs

Hi, my name is Nicholas and I am 9 years old. I have classic galactosemia. I like having galactosemia. My favorite foods to eat are eggs and bacon, eggs and ham, and pancakes. The thing I like at school is like everybody is all over me and saying what does it feel like to have galactosemia. The most fun thing is meeting people all over the world like me. I am counting on meeting all my new friends at the conference and to taste the foods. I hope I will have a good time.

My hobbies are baseball, basketball, gymnastics, playing playstation. I like to sleep late and stay up late. My favorite holiday is Christmas. The thing I want the most is a go-cart. My dad says I might be able to make a deal with him to let him pay half of it and let me pay for the other half. My favorite show is the suite life of Zack and Cody.

I have a friend that has diabetes I meet him in second grade. When I met him I thought I had a badder disease than him but really he had the most I was so happy to meet him. I thought that I was like the only kid in Mississippi that had a bad disease like what I have but that proves me wrong. When I was a little kid it was really hard for me to know what I could have to eat and what I couldn't.

This story is just about like what it is like what it feels like to have galactasemia. And it is about it is not that bad to have galactosemia.

Living with Galactosemia

Brett - California

My name is Brett, I am 18 years old. I was born in California and have classic Galactosemia. My parents found out I had Galactosemia when I was 6 days old. I was sick when I was a baby and my parents took me back to the hospital and they put me in ICU. I do not remember when I was sick. The first time I realized I had Galactosemia was when I started 1st grade and there was a pizza party for someone's birthday. I only had a tiny 4 slice pizza and the other kids could eat as much as they wanted. When there were cupcakes I would not be able to eat them, so if there was a kid's birthday and he didn't tell the class the day before, then I would not be able to eat anything. If they told us ahead of time, my mom would make a special desert for me like award winning brownies in the 2002 Galactosemia bake-off. She would make chocolate chip cookies or brownies for me and then I could bring my own dessert in. That was kind of hard because then all the kids would ask, "Why do you have your own"? They all wanted mine.

I can explain what Galactosemia is a little, but I probably don't understand all of it. I just tell people it is a rare, metabolic disorder and I don't have enough enzymes to digest the milk. I don't care to tell people that I have Galactosemia. When I tell people that I can't have dairy, how does it make me feel? Smart, because I get to explain how my body works. I just tell people that I can eat the same things as them; it just has to be specially made.

It is hard for me to do such things as write clear and complete thought sentences in English class, speak in clear and thought sentences so people understand me, my friends say I'm random. I will just say something out of the blue, random things off topic.

Do I feel different because I have Galactosemia; different from whom? That's what I want to know. I don't care that my mom or dad tells people about Galactosemia. It makes me feel more special than I am. Do you remember when you were little and what it was like to have Galactosemia? Yes, it was tough in a way because I knew I couldn't have foods although my friends and everyone else could eat whatever there was. But on the other hand, I am special getting my own custom pizza with whatever I wanted on it or cupcakes when everyone else had cake. Everyone would always ask why I got something different. They always wanted what I had, maybe because it was different. It is so hard to chose when you go out to eat because you have limited things to choose from. The hardest part about it is when you order something to remember to say no butter on the bread or no cheese on the salad because they do it automatically in restaurants.

Is it hard to find food that you can eat to order? It is generally easy for me to find food to eat. The servers are generally helpful when I tell them I cannot have dairy, but some people I

have talked to do not even know what dairy is and I have to explain it. Another thing that doesn't bother me too much, is when people at the table eat things I can't eat. I think it is because I'm used to it. All in all, I am pretty good about my diet and I don't think I am any different than the kid next to me.

Living with Galactosemia

Stacey - Canada

Galactosemia is not who I am. Who am I? I am a strong woman. I think outside of the box. I am more patient than others. I am accepting of who I am. I am not alone. I was diagnosed at six weeks of age; it was February 16th of 1977. If there had been newborn screening I may have been diagnosed sooner. My young, confused parents had to wait for a diagnosis while I remained a helpless baby, labeled as "failure to thrive", an enlarged liver, a dense cataract and clinging to life.

Doctors seemed puzzled as to why I was vomiting after feedings. It was suggested that perhaps I was lactose intolerant and would grow out of it over time. The lack of a galactosemic newborn screening test almost cost me my life; our family almost destroyed for a simple heel prick. I had been the first infant in ten years diagnosed with galactosemia at this hospital. I can't help but wonder how many babies did not make it within those ten years. As a parent it saddens me to think what my parents went through, watching their first-born deteriorate in front of them and being told it would be impossible for me to live a normal life because of the brain damage that had already occurred.

I was eventually sent home with a sample of soya milk and a piece of paper with an explanation of my condition, no more than a paragraph long. How perplexed my parents must have felt. During the years, I had a second cataract due to a galactose accumulation. School was the cause of immense frustration while growing up; I disliked school, I struggled to learn, I learned things at my own pace. I new knew then but I was a lonely child who never met another Galactosemic; in fact it felt as if I was the only person in the world with Galactosemia. I went through my early childhood years hating myself for being different, not understanding why I wasn't allowed to drink milk or have ice cream like my friends or why I was always the last to finish a simple math quiz. When I reached my teenaged years, I rebelled. I had numerous behavioral issues. I would often bump into things, my eyesight was poor, labeled by teachers as "clumsy", and my classmates had their own choice of names. I ate a large number of galactose and lactose containing foods not knowing the harm that consuming the "bad" foods was potentially causing.

I grew out of my teen years and into my mid adult life where I moved from my hometown to start college. Once I moved I met a man who would change my life from that point forward. His mother was a nurse and couldn't understand why my stomach would hurt after eating lactose containing foods; she found it terribly strange how I had no answers to why I couldn't eat certain foods. If it wasn't lactose intolerance, then what could it be?

The cataracts became more noticeable and it was becoming more of a hindrance. She recognized this and referred me to a geneticist at the local hospital. It was at that appointment that I learned to my disbelief that I was living with Galactosemia and what the disorder entailed. I had no clue. It was much more complex than not being able to eat certain foods. I am now on a stricter diet and since then I have had two cataract surgeries, my lens have now been removed and replaced with a plastic "foldable" lens.

My newfound eyesight has enabled me to learn again, to use my eyesight and not take it for granted. In a sense, I was reborn and I could now see the world with a new set of eyes. There was so much to learn, so much to take in, my brain was hungry and I could not digest the information quick enough. I for once in my life wanted to learn!! Life was not cruel, life was wonderful... beautiful! Since then I have made an incredible network of friends in the Galactosemic community, I have traveled many miles to attend conferences held by the PGC. Looking at it now, I don't think there is a limit as to how far I would travel to be with the people separated the deep rooted loneliness within me. For once, every two years my spirit is rekindled and I'm not just a statistic. It was at that genetics appointment that I discovered as a common side effect of Galactosemia Ovarian Failure has been documented in 85% of females. This would explain why I had very few menstrual cycles in my life, thus I was under the impression I would have premature ovarian failure. I remember going to doctor appointments to find out why I didn't have monthly cycles. The doctors ask me time and time again to explain to them what Galactosemia is. Feeling frustrated and lonely I again, did my own research for the doctors. Before seeing a fertility specialist, my husband and I never gave up hope, we welcomed any opportunity for parenting that came our way, conceiving our own child would have only been a bonus. We continued to hope. We hoped, we prayed, we fell, we cried, when we were told it couldn't be done, we rose to our feet again, continuing to hope, remaining optimistic. We tried for years, listening to the specialist's doubts, nodding their heads. I continued to hope for the best and keep my head up high.

I am happy to say that we now have a healthy two year old. I will teach my son that he is God sent and to honor his health and remind him that he is the face of hope for all. We are blessed by him and each day I look into his eyes and remind him that he truly is a miracle and my world has been made rich by him. I know he will give hope to those who too have doubted.

I do not want sympathy because I have galactosemia; rather I see it as an opportunity to provide optimism in the years to come for other galactosemics.

Living with Galactosemia

Keith- New York

My name is Keith. I am 9 y/o and was diagnosed with classic galactosemia when I was born.

Galactosemia is not a disease, illness, or sickness; I think it is a way of life. Galactosemia is an allergy. If you have galactosemia, you are allergic to cow milk, margarine, dairy, whey, and other cow milk products. Galactosemia is a big and important part of my life!

Just because my brother, Christopher Topper, age 8 and I have galactosemia it doesn't make us any different than kids who don't have galactosemia. I even think that he is one of the most popular kids in the 2nd grade and I am one of the most popular kids in the 3rd grade. We are both very smart and have lots of friends (even girls).

Galactosemia does not mean that we cannot accomplish things that we would like to accomplish or things that others can accomplish. My brother and I have received a lot of trophies from baseball, basketball, soccer and football. We have also received many medals for wrestling, chess, football, science fairs and the fire department drawing contest. We have accomplished many goals in different projects, sports and other events. We have earned many certificates. Some certificates are for Student of the Week, Science Fair Projects, being good in class and many, many, many more. Too many to count.

Though you cannot eat or drink food products that contain cow milk, in my opinion, Galactosemia is still not too bad. Galactosemia is fun whenever my brother and I go to birthday parties; our mom always bakes brownies or cupcakes just in case the other cake contains cow milk. We help my mom decorate the cupcakes and they always look very cool! All the kids eat them! Another fun thing about galactosemia is that whenever my family goes food shopping, my brother and I are responsible for checking the ingredients on the labels. We make sure they don't have dairy products. We are good at reading labels.

Whenever we go to a friend's house, my mom or dad call the parents to tell them about our allergy so they don't give foods that might make us sick. If we do eat a small amount of food or drink a little beverage that contains dairy, we might throw up and get very sick. We don't like that part.

Usually the food that we buy doesn't contain cow milk, but there are some things in our food pantries that do contain cow milk. Those foods and drinks are my mom and dad's. Whenever I

go into the refrigerator or the freezer I always know what I can and cannot have. If I don't know, I always check the ingredients. Some foods have ingredients that I am not familiar with and I always ask my parents.

Children that have galactosemia really shouldn't drink cow milk. If you can't drink cow milk, then what milk can you drink? That is the quest you were probably asking yourself. We drink soy milk. Milk that comes from cows is dairy milk. Dairy is not too healthy for galactosemic kids. Soy milk comes from soy beans and is healthy for us. I love chocolate soy milk.

My whole family, except for one member, knows about our allergy. Everyone, except for one understands what it means. The one who doesn't is my 2nd cousin and he just 10 months old. He has never heard of galactosemia and does not understand its meaning. That is o.k., he is only a baby.

In conclusion, galactosemia is not too terrible. The end.

Living with galactosemia
From a sibling's point of view

Tori - Mississippi

I am Tori, older sister of Landon and Tela. I am 10, Landon is 6 and Tela is 4. Landon is the only one with galactosemia out of the three of us.

My brother is the main reason we hardly go out to eat because he can't eat a lot of things. When we do go out to eat we have to bring a pack along that has his bread, butter and mayonnaise. He uses mayonnaise in place of ketchup. Landon usually gets his food last because his is a special order. So we wait and it sometimes makes me sad. It does not make me sad that we have to wait, just makes me sad that he gets his food last all of the time. If we go someplace where Landon can't have the bread or rolls, it makes me sad, so I don't eat the bread either.

I will sometimes order what Landon is eating just so he isn't the only one eating what he orders. Tela will do this sometimes too. We order plain noodles a lot because they are usually safe. Plain noodles just cooked in plain water and that is it. We like it too.

Just because he is galactosemic does not mean he cannot do certain things. Landon is very good at sports. He plays soccer, baseball, and swims well too. He played 1st base, shortstop, and pitcher this year and was good at all positions. He is excellent on his game boy too. Sometimes I get him to get me to the next level on my games.

Sometimes Landon having galactosemia does make me sad because he can't have a lot of things we eat. There are times when I wish I had galactosemia too. I have wished this so we could eat the same things and also so that Landon would not be the only one in the family with galactosemia.

I do wish a doctor could discover how he could eat more foods and not be harmful to him. If I had one wish for Landon, it would be for him to meet someone his age that he could be buddies with that was galactosemic too. That way he would not feel so different at school and different places we go. Maybe.

I love my brother very much. I think he is special because he is galactosemic. Mama says he is very special. I love having Landon for a brother. I would not trade him for another brother who wasn't galactosemic. I love him so much I wouldn't trade him for anything. He would not be Landon to me if he were not galactosemic.