

#### OFFICERS

President:  
Neal Stanger  
Vice-President:  
Lisa Weir  
Treasurer:  
David Wolfe  
Secretary:  
Brownie Shott

#### DIRECTORS

Cynthia Antaya  
Diane Haynes  
Matt Murray  
Marilyn Ogan  
Pamela Ryan  
Lori Swanson  
John Wynne

#### SPECIAL ADVISORS

Meg Hefner  
Kathy McNulty  
Susan Wolfe

#### ADMINISTRATIVE

ASSISTANT  
Lori Bookstaver

#### EDITORIAL STAFF

Lisa Weir  
Meg Hefner, M.S.  
Leslie Kauffman

#### INSIDE THIS ISSUE:

Introduction to the  
Special Issue

Family Stories

Prenatal Diagnosis of  
CHARGE

Diagnosis of CHARGE

Recurrence Risk

CHARGE Humor: New  
Takes on the Acronym

Bay Area Sisters and  
Brothers In CHARGE

Research Opportunity:  
CHARGE & sleep apnea

An Update on Gary  
James

Southeast Regional  
CHARGE/DB Weekend

# CHARGE Accounts



VOLUME 20, ISSUE 2

SUMMER, 2010

## Introduction to the Family Planning Special Issue



**By: Meg Hefner, M.S., Genetic Counselor**

Every so often either on the CHARGE syndrome listserv (<http://health.groups.yahoo.com/group/CHARGE/>) or in emails sent directly to me, people ask questions about how having CHARGE or having someone in the family with CHARGE (previous child, brother or sister, cousin, etc.) affects the possibility of CHARGE in a particular pregnancy. People ask me about the chance of another child with CHARGE, at what point it might be diagnosed and how, whether it would be “better or worse” than their first child, and what other people do. In addition to the enquiries that come directly to me are the discussions on the list about the worries, fear, guilt, and all the other feelings that accompany the process of deciding to attempt a pregnancy, the uncertainties during a pregnancy, the joy of birth and the tragedy of pregnancy losses. These are themes which reappear over and over again in our families. In this special issue of *CHARGE Accounts*, we will try to address many of those issues, providing you with technical medical information about probability and medical testing along with stories from families. This issue will become another resource the Foundation provides for families in the future.

Many people have contributed to this issue. Although I have written many of the more technical articles, my information comes from numerous professional sources, both in the CHARGE world and in my “other” world as a prenatal genetic counselor. Corrie Young writes about on her experiences with pregnancies after her first child with CHARGE, including several typical children and one more child with CHARGE. Others have contributed their experiences of deciding whether or not to pursue additional children after their child with CHARGE. The editors wish to thank all of those who contributed to this issue.

### Editorial comments from Meg:

This is an extremely important topic with some very strong feelings and opinions. As a genetic counselor, I work in a high risk prenatal genetic setting every day. One of the most important tenants of genetic counseling is to always be “pro choice.” Unlike what you might think based on what you see in the media, this does not mean “pro abortion” (or pro g-tube or pro tracheotomy or pro cochlear implant...). It means that every person, every family, deserves the right to make whatever decision (choice) is best for them. There are no right or wrong decisions, only the best people can do with the information they have. Doctors or legislators should not be the ones making these decisions. People are often thrust into situations where they must make heart-rending, irreversible decisions with incomplete information. My role is to help people understand a situation as completely as possible and help families see how different decisions might affect them and their families, now and in the future, with the hope that the family can make the best decision for them. Once a decision is made, it is my job to support the family in that decision. Never to judge.

In this issue, all of the families who decided to pursue another pregnancy have written that they would take whatever they got – CHARGE or not. With the relatively low recurrence risk, it is not surprising that the Youngs are the only family who actually faced the prospect of another child with CHARGE. Corrie has eloquently related how they felt about that, fears and all. I have had many private conversations (phone and email) with families who felt pretty strongly that it would not be right for their family to have another child with CHARGE and yet they desperately wanted another child. Those women often clearly stated they would likely terminate a pregnancy that was found to be affected. I mention this only because I know there are others out there who may also feel that way, and that is OK. I know the world of CHARGE is very accepting and I hope that opinions and decisions made by people to actively not have another baby with CHARGE will be as respected and admired as those who have decided to accept whatever happens.

# Bumps in the Road

**By: Catherine Rose**

## How to start an article that can potentially make people cry?

*“Like many families, our journey to having kids was filled with bumps in the road.”*

Like many families, our journey to having kids was filled with bumps in the road. I had always wanted two children close in age. I'm an engineer, so I like to do things very efficiently. And to our surprise, in our first pregnancy, we were expecting identical twin girls. I was monitored closely by high-risk pregnancy doctors in Boston and had ultrasounds every 2 weeks to check the growth of Alexis and Kaitlyn.

During the 20-week ultrasound, the doctors noticed a heart defect in Alexis's heart, so we had an amniocentesis performed on both babies. We thought waiting for the test results were the most difficult days of our lives. The tests came back negative for both babies, which gave us peace of mind to get through the rest of the pregnancy.

At 29 weeks, I was admitted to the local hospital with fatigue and dehydration. At that time, the nurses and doctors couldn't find Kaitlyn's heartbeat. I was completely operating in pregnancy brain, thinking, “They just aren't hunting hard enough.” My husband is very rational and had already put together the pieces. The ultrasound tech confirmed the dreadful news: “Kaitlyn has no heartbeat.” I was transported immediately to Boston for lung steroid shots for Alexis, in case she was going to be born at 29 weeks.

After several days of observation, Alexis showed she was a fighter and was staying put. I was ordered to strict bedrest and endured another 8 weeks of pregnancy carrying live Alexis and dead Kaitlyn. We had a c-section scheduled because Alexis became IUGR (intrauterine growth restricted). At 37 weeks, Alexis was born weighing 4 pounds.



**Sisters Jessica (left) and Alexis (right)**

Through Alexis's first year of life, we identified the following genetic anomalies: right side choanal atresia, cleft of soft palate, bilateral severe profound hearing loss, small right eye, low vision, heart defects (ASD and PPS murmur), fusions of the cervical spine, microcephaly, clenched hands, kidney problems (pseudohypoaldosteronism type II), abnormal brain spikes, and obstructive sleep apnea. In the NICU, the doctors felt that Alexis had Trisomy 18, which is incompatible with life—giving her less than a 10 percent chance that she would survive her first birthday. She tested negative for every test. She tested negative for CHARGE as well. She also tested negative for Cornelia de Lange. She was undiagnosed, a medical mystery, a scientific misfit. If a child is diagnosed with a genetic syndrome, doctors can check the parents and siblings. If a child is undiagnosed, then they don't have anything to search for.

## So knowing what we did and experiencing the challenges that we did, why did we have a second child?

I had a lot of time for reflection during bedrest and NICU days. We had expected twins. We had prepared for twins and the struggle of caring for two babies. I knew if I wanted to have two kids close in age that we couldn't be deterred by the complexity of Alexis's care. We had 20 doctors following Alexis. She received about 10 hours of Early Intervention each week. And then, we got pregnant with Jessica when Alexis was barely 8 months old.

*Continued on next page*

Continued from page 2

### Others would question us, “Aren’t you afraid that Jessica will have the same genetic issues?”

Our answer was, “If that happens, we know all of the RIGHT decisions and already have our rolodex of doctors.” We were resolute that Alexis could overcome all of her challenges and felt that if Jessica had those same challenges, we were well-equipped to handle them. However, we were still very afraid and concerned for her well-being.

I had CVS testing done at 10 weeks to check for any genetic defects. I breathed a sigh of relief after the 29<sup>th</sup> week (when we had lost Kaitlyn). I was followed by high-risk pregnancy doctors and delivered Jessica safely by c-section at 38 weeks. After the delivery, we were hypervigilant—no cleft palate and no clenched fists. We were discharged within 4 days and were sent home with a baby who was actually hungry at night (no g-tube feeds). It was a new experience!

Yes, it has been crazy. In the early days, Alexis and Jessica had many competing needs. Now, Alexis is 4 and Jessica is almost 3. In a lot of ways, Jessica is the big sister, achieving the milestones that Alexis struggles to reach. Alexis has an excellent role model—taking notice of Jessica’s movements, trying to do similar actions, and sharing toys together. And Jessica is more caring, more patient, and more sympathetic than many of her peers because she has Alexis. We wouldn’t trade them and their interactions for anything. We are blessed by our beautiful girls.

## Prenatal Diagnosis of CHARGE Syndrome

**By: Meg Hefner, M.S., Genetic Counselor**

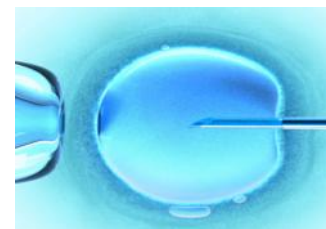
Can we look for CHARGE prenatally? What options are available depends on whether a CHD7 mutation (or other CHARGE gene –when we find them) has been identified or not. If a DNA diagnosis has been made, then all of the options below could be considered. CHD7 (or other DNA testing) can be important even when the clinical diagnosis is definite, because it means there are many more options available for family planning. If no DNA diagnosis has been made, skip to Ultrasound, below.

For any genetic condition, if the DNA basis of the condition is known, it is possible to test for the presence or absence of that particular mutation at many stages: before pregnancy (preimplantation genetic diagnosis), in the first trimesters (CVS), in the second trimester (amniocentesis), and postnatally. Prenatal diagnosis options should always involve extensive counseling about the advantages, risks and cost of procedures being considered. The option of terminating a pregnancy (abortion) varies from location to location – in many US states, it is not available after 22 or 23 weeks.

### Preimplantation genetic diagnosis

This involves IVF (in vitro fertilization) along with DNA testing of embryos prior to implantation. Briefly, hormone injections stimulate multiple ovulation. Eggs are harvested, fertilized with sperm, and allowed to grow to about the 8 cell stage. One cell is removed from each embryo and tested for presence or absence of the known mutation in the family. Only embryos without the mutation are implanted in the woman’s uterus. IVF with preimplantation genetic diagnosis is only done in a few centers. It is very expensive and does not always result in a viable pregnancy. The major advantage of this procedure is that it avoids the possibility of a decision about whether or not to continue an affected pregnancy.

*“Prenatal diagnosis options should always involve extensive counseling about the advantages, risks and cost of procedures being considered.”*



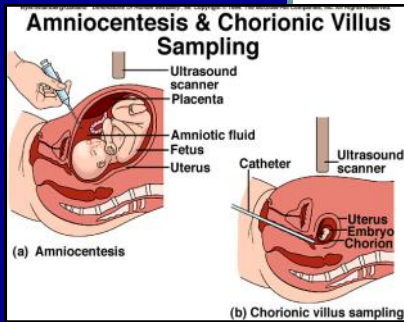
**Preimplantation genetic diagnosis involves in vitro fertilization and DNA testing.**

Photo credit: <http://www.scientificamerican.com/blog/60-second-science/index.cfm?tag=IVF>

Continued on next page...

## Prenatal Diagnosis—Continued from page 3

## Chorionic villus sampling: CVS



CVS is a prenatal diagnosis procedure done at about 11-13 weeks of pregnancy. A catheter is inserted vaginally or abdominally (depending on the position of the placenta) and placental tissue is removed. Fetal tissue in the placenta can be tested for presence or absence of the known mutation in the family. The major advantage of this procedure is getting information earlier: either the relief of an unaffected diagnosis or the ability to make a decision about continuation of pregnancy or abortion early in the pregnancy.

## Amniocentesis

Amniocentesis is a prenatal diagnosis procedure done after 15 weeks of pregnancy. A needle is inserted into the amniotic fluid surrounding the baby. A small amount of fluid is withdrawn. Fetal cells in the fluid can be analyzed for the DNA mutation in the family. The advantage of amniocentesis is the lower risk of miscarriage.

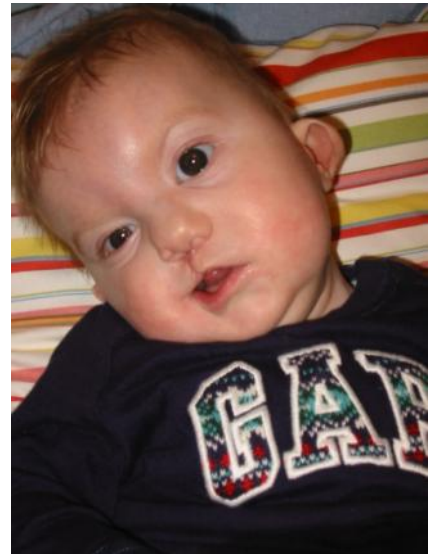
## Ultrasound

Ultrasound, unlike DNA diagnosis, cannot diagnose or rule out CHARGE syndrome in a pregnancy, even when the CHD7 mutation is known. However, ultrasound is an extremely useful tool and has no known risk to the fetus or the mother. An anatomic survey performed at a tertiary care center can provide a lot of information about the general well being of the fetus. We typically do an ultrasound for anatomic survey at 18-20 weeks. At that point, many fetal anomalies can be detected, yet it is still early enough in pregnancy to perform additional testing (e.g. amniocentesis to rule out a chromosome abnormality, fetal echo to look at the heart) and still have the option of considering pregnancy termination. When looking for features of CHARGE, I would recommend another ultrasound at 24-26 weeks, when a more complete anatomic survey (especially heart views) can be completed.

Ultrasound features which may be consistent with CHARGE syndrome are listed on a separate sheet (so you can print it and take it with you) on page 7 of this issue. A note about 3D/4D ultrasound: 3D ultrasound is really fun for parents. As a medical diagnostic tool, however, it is most often not as valuable as 2D ultrasound, Doppler (which looks at blood flow) and other things which don't look as cool to the parents. However, for certain features, 3D can be very helpful. In the case of CHARGE, getting a 3D look at both ears can be very helpful. Unusual ears are one of the most common features and can sometimes be very obvious on 3D ultrasound.



3 D ultrasound of Tate Young at about 24 weeks.  
Note the cleft lip and the CHARGE ear sticking out



Tate several months later - lip is fixed,  
ears still stick out

Continued on page 6



# “Four in Our Family, All Done”

**By: Katie Susil**

## THE DECISION

The decision to have another child after our first baby, Makenna, was born with CHARGE syndrome was not an easy one for us. In fact, Jay and I were positive we would not have any more children for the first three years of Makenna's life. Once Makenna's medical issues started to become more stable, I began to get that “itch” for another baby. It took a few more years and many, many discussions for Jay to get on board. What if the baby was born with CHARGE syndrome or something even more devastating? What if Makenna couldn't handle a sibling? What if the baby grows to resent Makenna for all the attention she requires? Is it fair to bring a baby into the world when we, as parents, know he could suffer as much as Makenna did as an infant? Could we live with that guilt? The list of “what-ifs” could have gone on forever. However, we finally decided that what will be, will be. If we were meant to have another baby with CHARGE syndrome, then we could handle it. After all, we had been through it before, and next time around we would at least know the ropes.



**Sisters Makenna (top) & Cami (bottom)**

## THE JOURNEY

Once we were expecting again, we had the hurdle of explaining to Makenna that a new baby was coming to join our family. She was in complete denial for a few weeks! She told me, “You don't have a baby in your belly. That is food, same as grandpa's belly.” When she came to terms with the idea, she told us it was a boy baby named Jacob. Needless to say, Makenna came to the ultrasound with us to find out the sex of the baby. I didn't want to be the one to break the news if Jacob turned out to be a girl. When the technician announced it was a girl, Makenna immediately turned to Jay and asked what her name was. We hadn't discussed that yet, so Makenna told us it was Cam, C-A-M, Cam.

Our trip to have a level-two ultrasound and genetic counseling was uneventful. The genetic counselor learned more from us than we did from her. The ultrasound looked normal; however, the doctor's parting words were, and I quote, “You never know—lightning can always strike twice.” Talk about NOT calming our nerves!

## THE AFTERMATH

Our little Camille, Cami for short, was born in November 2008 (seven years after our first child) as healthy as we had hoped! I thought I was ready to parent a healthy baby, but this was uncharted territory for us. It was such a strange feeling to take home a 2-day-old infant. She didn't have to pass a car seat test. There were no CPR classes or trach care classes or ventilator classes. There was no need to chart tube feedings, or dressing changes, or temperatures, or even count respirations. I looked at Jay and asked, “How are we supposed to know she is okay?” There wasn't a house full of nurses, emergency medics, and respiratory therapists when we came into our home. We were actually a little nervous to have a healthy baby. I may have even been a little overprotective. Cami slept with us for seven months because I didn't like not having heart monitors or pulse oxygen machines there to alert us if something was amiss.

Cami's birth has been great for Makenna. She has really grown in her self-awareness and independence. We have never hidden the fact that she has CHARGE syndrome from her; however, this made a nice transition to using the actual name as to why things are different for her than for Camille. She had, and continues to have, many questions about how Cami eats compared to how she did as a baby. She realizes not everyone spent time in hospitals and had surgeries as an infant. I think it opened her eyes to the fact that not everyone starts out sick. Makenna now wants to do things for herself and has grown much more independent in the last 17 months.

Cami is so very lucky to have Makenna as her big sister. She has already learned quite a bit of sign language and picks up new signs everyday. She loves Makenna so much. Cami always wants to know where Sissy is and what she is doing. Cami will grow up with compassion and understanding for people with special needs.

Of course, we still have challenges, but the rewards in having a second child have outweighed them by a huge margin. Jay and I are thankful everyday that we get to experience Cami's milestones with such awe and appreciation that we never would have had without our experience with Makenna. Each and every little milestone that Cami makes so easily is a huge miracle to us, and we get to experience that every single day. We have Makenna to thank for that.

Is our family complete? We haven't quite decided yet, but Makenna has her opinion. In her words, “Four in our family, all done.”

***“The variability of CHARGE makes decisions about family planning, prenatal diagnosis, and pregnancy management even more difficult.”***

#### Prenatal Diagnosis—Continued from page 4

Your fetus has CHARGE, what now?

Whenever I call someone with a positive prenatal diagnosis, the first question people always ask is “Are you sure (of the diagnosis)?” The answer is almost always “Yes, absolutely.” The second question is always, “Can you tell me how bad it will be?” The answer to that is almost always no. That is information people understandably want when making very difficult decisions about their pregnancy. Even with conditions such as Down syndrome, there is a wide range of both medical and developmental issues. In CHARGE syndrome, the range is even wider. And CHARGE does not necessarily “run true” in a family. In the families with more than one affected child, sometimes one died in the newborn period and the other is doing very well, or they have different features. Or a parent is only diagnosed after a diagnosis is made in their child.

The variability of CHARGE makes decisions about family planning, prenatal diagnosis, and pregnancy management even more difficult. If you knew what you were getting into, you could perhaps make a reasonable decision about it. The unknown is far more difficult. Ultrasound is the only thing that may be able to help a little bit during pregnancy. For example, one thing we know about CHARGE is that infants with a serious heart defect along with choanal atresia have the highest mortality rate. Many heart defects can be detected by a fetal echocardiogram. Choanal atresia in some cases can cause polyhydramnios – excess amniotic fluid, but often not until late in the pregnancy. Ultrasound findings might, in some cases, provide families with information beyond the DNA diagnosis. And for those without DNA testing, you could say, “Well, we don’t know if it is CHARGE or not, but at least the heart is normal.”

## A Different Path

**By: Lisa Weir**

When I married Graeme thirteen years ago, I had two sons from a previous marriage. Although we weren’t in too much of a hurry, we knew that we would like to add to our family at some point. We did end up going down the pregnancy path quite quickly though, when we learned we would be expecting our first child together early in 1998.

The pregnancy had been a very easy one, just as my other two had been. I seemed to be carrying more fluid this time but was not worried in the least. We had our typical, 18-week ultrasound and regularly scheduled doctor’s appointments, and nothing unusual was noted other than the increased amount of fluid. So we carried on, blissfully unaware of any complications whatsoever.

Kennedy arrived on January 27, 1998, and it’s a day none of us will soon forget. It was a shock to the system to learn that she was born with a malady I couldn’t even pronounce until several days later. Little did we know on that day, bilateral choanal atresia would be just a small part of what was to come. As many of the families will relate, though, things did eventually settle into a new routine of normalcy and we became comfortable with our new life on the CHARGE path. Hospitals, therapies, surgeries, and appointments became as standard as Monday night meatloaf.

Not a lot of time had passed when we began to think about future pregnancies, and we made the decision that we would not be adding to our family. I had felt strongly that I wanted to devote as much time as possible to Kennedy and her needs and that it would be difficult to do so with another child. I guess I felt that I wouldn’t be able to stretch myself to give both Kennedy and another baby everything they needed. I’d read many stories on the CHARGE list about families who had done it successfully and marveled at their ability and courage to do it again, but I knew it was not for me. Thankfully, Graeme agreed, and we made the decision that I would have a tubal ligation. My reasoning for wanting to be the one to have contraceptive surgery was that I wanted Graeme to have the opportunity to have more children should anything happen to me. I knew that after having had my own three children, I would be done, regardless of what the future would hold.

Needless to say, we certainly ended up on an alternate course than we had envisioned from the early days, but we are very happy and content on our different little path through life and with the decisions we’ve made.



**Top: Kennedy with brothers Kyle (left) and Ryan (right) in 1999**

**Bottom: Kennedy, May 2010**



# CHARGE Syndrome Ultrasound List

The ultrasound should be a **Level II ultrasound exam** and should be performed at a **tertiary care center** by an experienced ultrasound technologist using state-of-the-art equipment. This is not a procedure that can be done in the typical obstetrician's office. The ideal would be two ultrasound exams, the first at 18–20 weeks (post LMP – last menstrual period) and the second about a month later at 22–24 weeks. Take along the following checklist to give the sonographer the best information possible about what to look for.

## ULTRASOUND EXAM FOR FEATURES OF CHARGE SYNDROME: THIS WILL NOT DIAGNOSE OR RULE OUT CHARGE

The ultrasound evaluation should include a **complete standard anatomic survey** with particular attention to the following:

**Amniotic fluid measurement:** Look for polyhydramnios (excess amniotic fluid) associated (in late pregnancy) with choanal atresia, esophageal atresia, or poor swallowing.

**Cardiac evaluation:** CHARGE can include any heart defect, but the most common include tetralogy of Fallot with or without AV canal, and right-sided anomalies, including VSD and aortic arch anomalies. Many centers can do a fetal echocardiogram, but that is not indicated for low-risk patients with normal Level II ultrasound.

**Kidney:** Any kidney anomaly can be associated with CHARGE, including hydronephrosis (excess fluid in the kidneys), small or absent kidney, horseshoe kidney, posterior urethral valves, and cystic kidneys.

**Brain:** Dilated ventricles, absence of the corpus callosum, or any other structural abnormality of the brain—rare but not inconsistent with CHARGE.

**Face:** Cleft lip or cleft palate—3D/4D if there is any question of a cleft.

**Ear:** Abnormal shape or placement (take along a photo of CHARGE ears). CHARGE ears tend to be short and wide with no lobe and stick out from the head. \*\*3D/4D views of both ears are recommended.

**Genitalia:** Small penis in a known male fetus.

**Many abnormal findings would not be present early on and/or would be undetectable until later in pregnancy.** Don't be shy about asking how confident the sonographer is about the accuracy and completeness of the ultrasound exam. Every exam is different, and none will detect every birth defect during pregnancy. The accuracy will depend on a number of things, including how far along you are, the position of the baby, your weight (the image is not as clear when it has to travel through a lot of maternal tissue before it reaches the fetus), the quality of the equipment, and the expertise of the sonographer. What can be seen one day may not be visible another day.

Remember, with the possible exception of the ear, **NONE** of the major diagnostic criteria for CHARGE (coloboma, choanal atresia, cranial nerve abnormalities, hearing loss) can be definitively diagnosed by prenatal ultrasound exam. Although finding evidence of some problem or potential problem through the ultrasound exam would certainly raise the suspicion of CHARGE and can help parents be prepared for that possibility, it is not diagnostic.

And remember, **a completely normal ultrasound exam cannot rule out CHARGE.**

# Diagnosis of CHARGE Syndrome

By: Meg Hefner



## Clinical diagnosis

The gold standard for determining if a child has CHARGE or not is still clinical: based on the medical findings in the child, as evaluated by a medical geneticist. For a complete discussion of the clinical diagnosis, see the About CHARGE tab at the CHARGE Syndrome Foundation website ([www.chargesyndrome.org](http://www.chargesyndrome.org)) and/or the GeneReview on CHARGE syndrome at [www.genetests.org](http://www.genetests.org). The clinical diagnostic features (major and minor diagnostic criteria) are mostly conditions that can be evaluated in the newborn period, as that is when it is best to make a diagnosis, if possible. (Note that we stopped using the c-h-a-r-g-e features for diagnosis when the diagnostic criteria were first changed in 1995.)

## DNA (CHD7) diagnosis

DNA diagnosis involves testing for mutations in the CHD7 gene on chromosome #8. If the clinical diagnosis is uncertain, CHD7 testing can often be helpful. If a CHD7 mutation is found, the diagnosis of CHARGE is confirmed. If CHD7 is negative and the clinical diagnosis is only probable or possible (not definite), then some uncertainty remains about the diagnosis. If the clinical diagnosis is definite, CHD7 testing is still useful because it confirms the diagnosis beyond any doubt and often provides information for family planning. In 2009, 10–30% of children with a definite clinical diagnosis did not have identifiable CHD7 mutations. Negative CHD7 testing does NOT rule out CHARGE syndrome.

## Why can't we find CHD7 mutations in everyone tested?

There are several possibilities:

1. Some probably have a mutation in CHD7 that is difficult to detect with the probes available now (2010). Retesting in the future might identify those.
2. There may be one or more other genes that cause CHARGE syndrome. Researchers are looking for those genes.
3. Some may have other conditions that have many overlapping features with CHARGE. Some of the conditions that can look a lot like CHARGE are listed in the Differential Diagnosis table. Most children with possible CHARGE had chromosomes and FISH for 22q deletion (diGeorge) as part of their workup to rule out those conditions. As children grow older, they may "grow out of" the clinical diagnosis of CHARGE and "grow into" features that give us clues to other syndromes, such as kabuki syndrome.

Those without a definitive diagnosis should return to a medical geneticist periodically (every 2–3 years) for reevaluation.

"The gold standard for determining if a child has CHARGE or not is still clinical..."



## CHARGE Accounts Welcomes New Editor

I would like to take a moment to welcome Leslie Kauffman to the *CHARGE Accounts* team. In addition to being mom to adorable Miss Katie and wife to Arlin, Leslie is a textbook editor with many years of experience. We are thrilled that she has agreed to come on board to help with the editing duties here at *CHARGE Accounts*. Her expertise is greatly welcome, and we appreciate her volunteering to become a part of the team.

Lisa Weir



# Surprise Baby

**By: Lori Myers**

I have three girls. I had two miscarriages between my first child and Sarah, my daughter with CHARGE, and the thought of another possible heartbreak was overwhelming. However, I was lucky and didn't have to choose to have another child. Thankfully, I had a "surprise" baby after Sarah.

I must say that my third daughter, Emma, completes our family, and I cannot imagine our lives without her. I was in awe of her health and how easy it was for her to thrive, and I was so appreciative and re-

spectful of this. I am so happy that she is here and that we all got to experience the miracle of a healthy child.

I am also thankful that Rachel, our oldest, got to experience the healthy birth of a sibling as well. Rachel was 3 when Sarah was born, and it was very scary and traumatic for her, too. Her life also changed. We gave Rachel naming rights to our third child, and after we vetoed "Pocahontas," she chose "Emma."

Now that they are all older, I am amazed at how they all take care of

and love Sarah. Not to mention, Sarah is Rachel's and Emma's biggest fan!



**Sisters (L-R) Emma, Sarah, and Rachel**

## Hard Decisions

**By: Leslie Kauffman**

My husband Arlin and I waited 11 years after we were married to decide to have kids. When we finally decided to expand our family, it didn't turn out to be an easy process for us. It took a year for me to finally get pregnant, only to suffer a miscarriage in April of 1999, followed by another in December 1999 and another in December of 2000. We were beginning to wonder if we were being given a sign that having a baby just wasn't meant to be for us.

In April of 2001, I became pregnant again, and this time we were able to sustain the pregnancy through the first trimester under the care of a high-risk pregnancy obstetrician. However, our joy was temporarily shattered in early August when, through a level-two ultrasound, it was discovered that our baby had a cleft lip and a heart defect, among other possible complications. We were so scared and heartbroken, yet this news only increased our love and compassion for our spunky little unborn girl, who proved her feistiness everyday with her nonstop energy and movement.

Katelyn Anne Kauffman was born on Thursday, January 17, 2002. While she did have a cleft lip and palate as shown on the ultrasound, her heart defect turned out to be minor and not in need of immediate treatment. Though there were some initial concerns about her kidneys, those checked out fine, and she was able to come home after only 48 hours in the NICU—in hindsight, a rare accomplishment in the world of CHARGE syndrome.

Although we were home, Katie struggled to eat by mouth, having to be fed every two hours with a special bottle, and usually taking 45 minutes to an hour to take 2 ounces of formula. Feeding was practically nonstop, and weight gain was very slow—but yet was so critical for getting her to a weight where she could sustain the many surgeries needed to repair her severe cleft. In between feeds, Katie was a very happy and alert baby. I remember holding her in my arms numerous times and thinking what a miracle she was.

Katie failed her newborn hearing screening, but they really couldn't get reliable results from the test since she was crying the whole time. So we repeated a series of hearing tests when she was a month old and found out she was severely to profoundly deaf in both ears. This was devastating news to us. I especially took this news hard, wondering how I was going to communicate with my child and how I was going to share my world with her. The following month, we had her eyes tested because

*Continued on next page...*

*"...she was able to come home after only 48 hours in the NICU—in hindsight, a rare accomplishment in the world of CHARGE syndrome."*

Continued from page 9



Leslie, Katie & Arlin  
Kauffman

***“We decided  
that our  
family was  
complete with  
just the three  
of us.”***

her left eye appeared smaller than the right. We learned that she had colobomas in both eyes, with the one in her left being in the optic nerve, making her completely blind in that eye except for possibly detecting light and shapes. Shortly after that, Katie had a CT scan of her ears, and we found out she was completely missing her semicircular canals, which control your balance, and thus would likely have delayed gross motor skills, including walking. Again, we were devastated. How could all these challenges be happening to our sweet little girl?

It was the combination of all these diagnoses that put together the puzzle of CHARGE syndrome for our geneticist. Fast forward eight years, and we have overcome nine surgeries and countless doctors' appointments and therapy sessions. Katie is progressing slowly but quite well in our public school system with an interpreter and one-on-one aide. She still has significant delays, particularly in expressive language, processing, social skills, and some motor skills, but she continues to amaze us every day with her abilities, quick memory, and happy nature.

During the past eight years, Arlin and I considered at times what it might be like to add another child to our family. Unfortunately, two years after Katie was born, I was diagnosed with a prolapsed uterus and had to have a hysterectomy, so additional biological children turned out to be out of the question for us. We did discuss adoption a few times, but eventually decided against it. Among many other things, our ages were a big concern. Given that we waited later in life to have Katie, I would have been approaching 50 by the time an adoption would have gone through, and Arlin would have been in his early 50s. Although we knew we could offer a warm and loving home to another child, we just weren't sure we wanted to give that child the ultimate eventual responsibility of not only being involved in Katie's care as we aged, but also having to possibly be our own caretakers at a very young age. It was a hard decision for us, but we decided that our family was complete with just the three of us.

I admire those who go on to have other children, either biologically or through adoption. Children are beautiful miracles who bring everyday joy to our lives. I also admire those who make the decision not to have more children, as I know how hard it is to make a decision that others may disagree with, but that is the right one for your own family. One child or ten, I think all parents of children with special needs deserve a huge pat on the back for the difficult decisions we make on a daily basis to ensure the best life for our kids and our families.



### **CHARGE Humor: New Takes on the Old Acronym**

Recently on the CHARGE Listserv, Desiree Massie challenged us to have some fun recreating the CHARGE acronym to reflect the, “wonderful, chaotic world of CHARGE syndrome”. And, since the acronym is no longer used in making the diagnosis, we might as well have some fun with it!

#### **Desiree Massie**

C - Chaotic  
H - Hospitals Galore  
(Groan!)  
A - Annoying  
R - Really Unfair  
G - Groan! Not another  
Surgery!  
E - Extremely crappy

#### **Chip Dixon**

C: Cool  
H: Humorous  
A: Amazing  
R: Rare  
G: Go Getter  
E: Energetic

#### **Ellen Howe**

C: Crazy  
H: Happy  
A: Amazing  
R: Real complicated  
G: Great  
E: Entertaining

#### **Keith Persico**

C: Challenging  
H: Hassle, and an inconvenience  
A: Annoying, especially when it gets  
in the way  
R: Real pain in the rear  
G: Growth problems? Really?  
E: Egads, what's with all the doctor  
appointments! ???

#### **Lisa Weir**

C- Courageous  
H- Humorous  
A - Adaptable  
R - Resilient  
G - Gregarious  
E - Effervescent

# Recurrence Risk in CHARGE Syndrome

For families with one child with CHARGE, the recurrence risk for CHARGE is about 1-2%. This is an “empiric” risk - based on experience and on the fact that CHD7 is a dominant gene. The overwhelming majority of kids with CHARGE are the first one in the family with the gene. In a small number of families, a parent also has CHARGE (and carries the gene). And in a small number of families, one of the parents has additional sperm or eggs which carry the CHARGE gene. When that happens, it is called “gonadal mosaicism.” In other words, some of the gonads (eggs or sperm) carry the mutation and others don’t, making a “mosaic.” In families with more than one child with CHARGE, the chance for CHARGE happening a third time could be as high as 50%.

Possibility of CHARGE syndrome in a pregnancy

| IF YOU ARE                              | YOUR CHANCE | BASED ON                                      |
|---|-------------|---|
| *Someone in the general population      | 1/8000      | Birth incidence                               |
| *A parent with one child with CHARGE    | 1-2%        | Empiric recurrence risk**                     |
| *A parent with two children with CHARGE | 50%         | Gonadal mosaicism                             |
| *A sibling of an individual with CHARGE | 1/8000      | Birth incidence                               |
| * Someone with a relative with CHARGE   | 1/8000      | Birth incidence                               |
| Individual with CHARGE                  | 50%         | Autosomal dominant (AD) gene                  |
| Two parents with CHARGE                 | 66%         | AD, and assuming two CHD7 mutations is lethal |

\*with no features of CHARGE syndrome and/or has been shown to not carry the CHD7 mutation in the family. If any family member has features of CHARGE, please consult a medical geneticist about CHD7 testing or other evaluations to look for a possible “mild” case of CHARGE.

\*\*The risk for each family is in reality either very low or very high, but as we cannot tell the difference between these families, we use 1-2%, which is based on observing what has happened in thousands of pregnancies.

## The Youngs: Experience Times Two

By Corrie Young

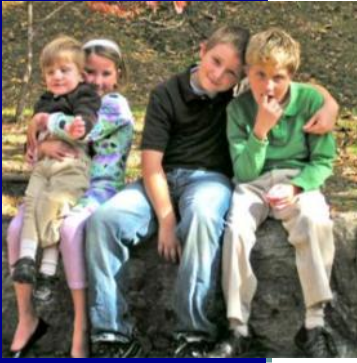
Almost 14 years ago, our lives became forever changed with the birth of our first child. Our son, Peyton, was born with CHARGE syndrome. Many of you know these feelings and can relate. As Peyton grew and developed slowly, according to his own timeline, we wished for a sibling for him. We actually wished he could have an older sibling to help engage him and become another communication partner in his little bubble of a world created by deafblindness. This was obviously impossible, so we prayed for another baby and were blessed a little more than three years later with our second son, Cy, born without CHARGE.

For my husband Mike and me, deciding to have future children was never a question for us. We took the odds of a 1–2% recurrence risk and ran with the 98–99% chance that all our future children would be born healthy and typical. So three years later, we were blessed once again with the birth of a baby girl, Mary Catherine. Our family seemed complete, but we were always open to future life. When our daughter was almost 3½, we learned we were expecting again. The news was both exciting and overwhelming, knowing how full our lives were with a son with CHARGE and two healthy, active, very young other children.



**Peyton Young**

*Continued on next page...*



Tate, Mary Catherine,  
Cy and Peyton Young

*“...as each week  
went by, we  
gained more and  
more confidence  
that the baby  
would be healthy,  
along with  
acceptance if the  
baby did have  
CHARGE.”*

We tried to prepare for the increased activity another baby would bring, but we never suspected what we would find at our level-two ultrasound. We went in with a bit of apprehension due to past experience, but having two typical children since Peyton gave us great confidence that we would see a healthy baby. However, this was not the case, and we actually saw more severe defects than those with which our first son was born. Peyton’s genetic defect was not yet found at the time of our fourth baby’s ultrasound, so we couldn’t “look” for that in him to confirm CHARGE. However, we immediately knew in our hearts it was.

We did all the genetic testing available at the time (ultrasounds, amniocentesis for chromosome analysis and 22q deletion, CMA), but all it did for us was rule out other syndromes for which it was able to test. A simple 3-D ultrasound in our doctor’s office here in our hometown, not two hours away where we were being followed by perinatologists, confirmed CHARGE. One look at our baby’s ears and we instantly knew he had this syndrome. We were followed, and our baby’s progress was closely monitored, particularly by cardiology due to the severity of his heart defect. Almost ten years after our first encounter with CHARGE, our fourth child, our son Tate, was born. We were once again brought back to the chaotic, frightening days of infancy with CHARGE.

This was an extremely difficult time for us because Tate was a more medically severe case and we had many more scares with him. His first surgery was at 2 days old, and his first open heart surgery would follow in another two days. I often wondered if he would have made it if we hadn’t had Peyton to prepare us. There were 911 calls by his older brother while I administered emergency CPR in the car as Tate lay limp and gray in my arms; rides in the ambulance to our Children’s Hospital two hours away; and unscheduled emergency heart surgeries (two), where Mike and I waited hours just to hear if Tate would pull through. In many ways, Peyton was such a blessing to his baby brother by giving us ten years’ experience under our belt of being parents of a child with CHARGE.

Fast-forward almost four years now, and Tate is an absolute joy in our family. Gone are the days filled with fear and caution about him just breathing and living. Yes, there are the many other challenges faced with having a child with CHARGE, but all parents of these special children know these challenges are far outweighed by the happiness these kids bring to our lives. I honestly can say, as I hope many of you can, that I wouldn’t change a thing about Tate or the fact that he has CHARGE.

With the birth of a second child with CHARGE, we were placed in a unique group of people who are thought to have *gonadal or germline mosaicism*. What this means is, my husband and I don’t carry the mutation in our blood, but one of us has the mutation in our sex cells that make sperm or eggs. (They did find our boys’ CHD7 mutation after Tate was born.) Not being able to test for our mutation, doctors gave us the possibility of having as high as a 50% chance of having future kids with CHARGE. We felt a bit daunted by those odds (are you a “glass half full or empty” kind of person, especially with our track record?). However, as practicing Catholics, we don’t use artificial contraception. Remaining open to life, we honestly in our heart of hearts hoped for our family to be complete, but that was not in God’s plans.



The Youngs in 2007

The beginning of November 2009 brought the news that we would be blessed with another baby. Being too early to identify CHARGE, we were very scared. Actually, that’s putting it mildly—we were beside ourselves! But as each week went by, we gained more and more confidence that the baby would be healthy, along with acceptance if the baby did have CHARGE. I know that may sound contradictory, but it can only be explained as a grace we received through lots of prayer. (If you’re a person of faith, you know there’s nothing like the CHARGE experience to get you praying! ))

In late February of this year, an excruciatingly long 16 weeks after we found out we were expecting (nothing like the CHARGE experience to teach you patience, either!), we peeked at our baby and found everything to look good. This time around, we elected to decline amniocentesis. We went back for follow-ups four weeks later and again ten weeks later. Both times we found the baby still looking good. We are praying to be blessed with a healthy baby girl in mid-July.

For us, in the beginning, questioning whether we would have more children after the birth of a child with CHARGE was easily answered. Yes, our faith has been tested as our family grows, but as many of you can relate, opening yourself up to the world of CHARGE is a life experience that I wouldn’t wish on anyone, but feel completely blessed to be a part of. (Am I contradicting myself again?) 😊



# CHARGE Syndrome Differential Diagnosis Table

| Feature  | CHARGE              | VCF/del22q          | Kabuki                      | VACTERL      | PAX2     |
|--|---------------------|---------------------|-----------------------------|--------------|----------|
| Coloboma   | +++                 | +                   | +                           | +            | +++      |
| Choanal atresia  | +++                 | +                   | +                           | -            | -        |
| Cranial nerves<br>Swallowing   | +++ long-lasting    | + short term        | ++                          | -            | -        |
| Smell  | +++                 | -                   | -                           | -            | -        |
| Facial palsy   | +++                 | +                   | -                           | -            | -        |
| Outer ear  | +++ short, square   | ++ long, narrow     | + big lobe                  | -            | -        |
| Hearing loss   | +++ mixed           | +conductive         | ++                          | -            | +++      |
| Cochlea abnormal   | +++                 | +                   | ++                          | -            | -        |
| Vestibular dysfunction   | +++                 | -                   | +                           | -            | -        |
| Cleft lip/ palate  | ++                  | +++                 | ++                          | +            | -        |
| TE Fistula or EA   | +                   | +                   | -                           | +++          | -        |
| Heart defect   | +++                 | +++                 | ++                          | ++           | -        |
| Kidney problems  | ++                  | +                   | +                           | +++          | +++      |
| Limb anomalies   | +                   | +                   | ?                           | +++          | -        |
| Genital/puberty  | +++ late            | +                   | + early                     | +            | -        |
| Behavior   | +++ OCD, other      | +++LD, psychiatric  | ++ sensory integra-<br>tion | -            | -        |
| Features   | Square face + hands | Long face + hands   | Unusual eyelashes           | -            | -        |
| Other  | Many other features |                     | Fingertip pads              | Anal atresia | -        |
| Diagnostic test  | Clinical, CHD7      | FISH for del22q11.2 | Clinical                    | Clinical     | PAX2 DNA |
| +++ very common    ++ occasional    + rare    - not seen    ? not sure |                     |                     |                             |              |          |

## Twins after CHARGE

**By: Kim Clayton**

When my husband and I got married, we said that we wanted to have three children. I even had the years picked out for when I would have those children. The timing did not quite go according to my plan, but in the end, I had not only three, but four, children!

When I was in labor with our second child, Christopher, we were asked if we wanted my tubes tied afterwards. My husband and I looked at each other, and we both said no. Before going into labor with Christopher, my husband and I had said we were finished having children after him because of various reasons—one being that we had put 10 years in between our first child, Johnathan, and our second child. We had no idea at the time that anything was going on with Christopher. Something in our hearts just told us that we would have more children, and that is why we said no.

*Continued on next page...*



**The Clayton Boys:  
Tyler, Cameron,  
Johnathan (standing)  
and Christopher**

*"We never really  
thought twice  
about having  
another child—or,  
as it turned out in  
our case, two!"*

Christopher came to us early and put us through a roller coaster ride for the next 72 days in the hospital. We didn't know during those 72 days that Christopher had CHARGE syndrome. We didn't learn that Christopher had CHARGE until he was 10 months old. Christopher had heart surgery when he was 5 days old due to Tetralogy of Fallot; he has bilateral colobomas, has profound hearing loss in his right ear and moderate to severe loss in his left, is g-tube fed due to major reflux issues, and is very orally defensive. He is also very small for his age and is developmentally delayed as well. He has never been tested for the gene—CHARGE wasn't genetically linked when he was diagnosed.

When Christopher was around 3 years old, my husband seemed to get "baby fever." I was really surprised to hear my husband tell me he wanted another child. The reason I was surprised is not because Christopher has CHARGE, but because my husband really wanted to wait until we could afford another child after our first one. Of course, I got baby fever right along with him (I probably had it secretly before he said anything), and we decided to try and have another child. We weren't worried or scared about having another child with or without medical complications.

We really shocked everyone when we gave them the news that we were pregnant with our third child. We were really shocked when we were given the news that we were pregnant with two! That is why we call Cameron our "bonus baby." We had to see the high-risk obstetrician

throughout the whole pregnancy because of being pregnant with twins and because we had a child with CHARGE. When asked the question, "What will you do if you have another child with CHARGE?," my answer was always, "Take care of the child just like I have taken care of Christopher."

The twins were born, and they are perfectly healthy! Are we done now? Yes, is the answer. I would love to adopt a child that has special needs. That, at this moment, is only a dream, a dream that may never come true. Some people think I'm crazy when I tell them I would love to adopt a child with special needs, but that's OK. I feel that Tyler and Cameron have been a blessing on our family and on Christopher.

You can tell that Christopher wants to learn right along with the twins, and I believe that they have helped him grow, learn, and thrive even more! Christopher loves to antagonize his brothers just like a typical big brother would. I was also always concerned that because we put 10 years between our first and second children, they wouldn't have a relationship. However, they share a very special bond together. Christopher thinks that Johnathan hung the moon, and Christopher is Johnathan's hero!

I would say one of the only challenges we have had with having another child after Christopher was taking the twins to doctor visits with Christopher when they were little and trying to juggle all three of them together. One of the challenges now is to explain to the 3-year-olds that they cannot do certain things that Christopher does, but that happens within a family with all typical children too!

We knew having another child after Christopher was going to be an interesting journey just because of all of Christopher's needs, but we also knew that we were going to be able to take care of another child no matter what was thrown our way. We never really thought twice about having another child—or, as it turned out in our case, two! It has been a wonderful and sometimes challenging journey. We have four wonderful sons, the three little guys are growing and thriving together, and we have been very blessed with it all!

## Conceiving Again

**By: Sandy Kurby**

I had twin boys, one (Joshua) born with CHARGE. He was a fighter. After 8.5 months, he completely blindsided all of us and moved on to Heaven. The doctors, my husband, and I were dumbfounded. He died of reasons outside of CHARGE (they called it an abdominal catastrophe since they really had no explanation). I had tried for years to get pregnant with my boys and finally had done IVF. I was on bedrest at 19 weeks, and delivered Joshua and Joseph at 32 weeks and 6 days.



**Joshua and  
Sandy  
Kurbly**

*Continued on next page...*



Continued from page 14

**Jillian  
Grace  
Kurby with  
mom,  
Sandy.**

As most of you can relate, our days had been filled with many therapies, doctors' appointments, and, of course, hospitalizations and surgeries. We wondered if financially we could afford to have another child, and if physically and emotionally we'd be available for a third child. We were back and forth on if we could afford (time- and moneywise) to have another child. By the time Josh was 6 months old, he was stabilizing at home, we were in our routine, and his brother was doing well too. We felt that we could in fact have another child and were not going to close the door to that possibility. As God wanted, Josh was admitted to the hospital for a month and a half and at 8½ months passed away (March 14, 2009). We were completely devastated. We didn't know how we'd continue without him here.

My husband and I decided shortly before Joshua's passing that we would try for another child, eventually. After he passed, I was still pumping/breastfeeding his twin so never got a period, but a few weeks later felt the need to take a pregnancy test (never had maternal instinct before!). I never would have thought it would be positive because of all the complications I had trying to get and stay pregnant previously. Well, early April we found out I was pregnant. While we were not preventing a pregnancy or ruling out another child, we had such a hard time conceiving and staying pregnant in the first place that we were in disbelief. Don't get

me wrong—we were very happy, but in shock. In telling people that we were pregnant after the first trimester, we heard many different attitudes. Some people were ecstatic for us; others questioned if we were trying to "replace" Josh, which was and is not the case.

During the pregnancy we wondered: *What if this child has CHARGE as well?* Many conversations with my husband resulted in the same ending: we would deal with whatever came our way. We loved Josh so much and would embrace having another child like him; we knew we would make things work with whatever was brought our way. There were times during the pregnancy that I wondered if she'd have CHARGE (and to be honest, after my journey with Josh, there have been and still are times I wish she did have CHARGE!). My OB sent me back to the perinatologist to be closely monitored. We had several growth ultrasounds, a very intense 20-week ultrasound, and as a reassurance we also had an echocardiogram done on the baby by Josh's cardiologist (who knew what Josh and his heart were like), but that showed nothing of concern. We were told that although the ultrasound and echo showed nothing, there was still the possibility of CHARGE. My OB encouraged me to have a quad screen test; however, I still decided not to.

All turned out fine. Jillian Grace was born at 39 weeks (c-section) and is very healthy. She had an Apgar score of 10, whereas we never got the Apgar score for Josh. The whole birthing experience was dramatically different. I was able to hold my baby this time, see my baby, go to recovery with her, and feed her. I remember as they were pulling her out, holding my breath just waiting to hear her cry, to know she was breathing on her own. When she was brought to me, the first thing I did was look at her ears. I will say, I was a little disappointed I didn't see those floppy ears that I loved to kiss on Josh. And our first question for the pediatrician once we were back in the room was, "When will she have her hearing screening?"

During her second day of life, Jillian choked on some fluid and turned a little blue. We had to hit the call button, and I had to run down the hall for help. They did the Heimlich Maneuver on her and then suctioned her mouth out. It was a good scare, but we had been through much worse with Josh. It was, however, a reminder of how precious and fragile life is. We are very glad that we did decide to have another child. We miss Josh dearly and he is still a part of our daily conversations, and always in our heads and hearts, and we are glad that he is Jillian's guardian angel and has sent her to us. Jillian and Joey are blessed to have an angel always looking out for them.

Jillian is now 4 months old and is very happy and healthy (outside of a cold here and there). She is gaining weight so fast it is amazing—she is going to catch her 21-month-old surviving brother soon in the weight department! I am so glad we did have her—we are truly blessed. Joshua taught us so much about love and life: that material things are not important; it's the smiles, the little accomplishments that some may take for granted that mean so much; and that life is precious. I have learned so much from Joshua and am so glad he introduced me to this wonderful community. I stay involved by reading (and replying when I have something to share) the CHARGE listserv, keeping a CHARGE Accounts fundraising page, reading and keeping up with many CHARGE kiddos' blogs (I'm really addicted!), and hosting a yearly 5K walk/run in memory of Joshua with all funds raised going to the CHARGE Syndrome Foundation.

**"...we knew we  
would make  
things work  
with whatever  
was brought  
our way."**

**2nd Annual Joshua Kurby Charge for CHARGE 5K—Saturday, August 14, 2010, Armstrong Park, Carol Stream, IL. We hope you join us for this fun day of walking, running, raffles, and more. If you are not able to make it this year, but would like to donate please visit this link to do so: <http://tiny.cc/kurbyrun> . For more information or to register, please email Sandy Kurby at [Sandy417@hotmail.com](mailto:Sandy417@hotmail.com)**

## BASIC: Bay Area Sisters & Brothers in CHARGE



**Top row:** Moriah Bettencourt, Danya Hardwick, Tiffani Lanting, Hannah Manniago, David Brown, Lacey Friedman, Nikki Harden  
**Bottom row:** Victoria Criswell, Victoria Nelson, Tanya Rago  
 (Missing from photo: Franca Angeli)

**BASIC has  
 now  
 blossomed  
 and hopes to  
 have 20  
 families in  
 attendance  
 for a summer  
 event!**

Last winter, three CHARGE moms decided to meet near Stanford Hospital in Palo Alto, California, for lunch. When the meeting date arrived, the group had grown to five CHARGE moms. Via word of mouth, the next lunch grew to 11 moms! Organizers (and mothers of children with CHARGE) Lacey Friedman and Victoria Criswell sensed the San Francisco Bay Area could use a support group not just for moms, but for all family members, of children with CHARGE syndrome.

A name for the group has now been coined: BASIC, for the Bay Area Sisters and Brothers In Charge. The name BASIC reflects many things, including learning to enjoy the basics in life as well as reminding us that we are not always in "charge" of our destiny (but nonetheless we continue to have hope and faith). BASIC has now blossomed and hopes to have 20 families in attendance for a summer event! The first two meetings included

heartwarming introductions, tons of pictures of our little ones being passed around, and information sharing. However, the group hopes to eventually branch out to offer outreach services for new families to area hospitals.

At the most recent meeting, BASIC was fortunate enough to have David Brown from California Deaf-Blind Services as a guest. David came away from the meeting refreshed, noting, "It [your group] had a profound impact on me. Very instructive and very healthy for me to hear you all remembering so much. And it has made me even more amazed that every one of these kids, even the most severely challenged, comes out with bags of strong personality, a bloody-minded determination to engage with the world on their terms...and as I looked around your circle on Sunday afternoon, what do you think happened? I failed to find a single shy, mild-mannered, poorly informed, hesitant, unassertive, smaller-than-life mom...all this horrible stuff certainly helps (makes?) you grow in extremely positive and powerful ways. Whatever you each got personally from the meeting, please believe me that you also helped me to take your inspirational behavior and use it on a much wider scale."

Knowing that there is very little that is "BASIC" about CHARGE syndrome, the group hopes to reach out to as many new families as possible. Current BASIC members are already reaping the benefits of having new friends who completely understand our complex lives and who can share information about doctors and therapists, medical tests, insurance, symptoms, educational rights, schools, and many other aspects.

On June 5, BASIC met at the California Deaf-Blind Services picnic in Hayward, California, where the group got its first chance to meet each other's precious children! Lacey and Victoria will host a separate family meeting in late July. For more information or to attend one of the planned summer events, please email [Lacedoxie@yahoo.com](mailto:Lacedoxie@yahoo.com) or [billvictoria@sbcglobal.net](mailto:billvictoria@sbcglobal.net).

## Wisconsin Dells Weekend Canceled

Unfortunately, due to some health issues, the weekend scheduled for this summer at the Wisconsin Dells has been canceled. Organizer Crystal Masionis has said she will try to get the event going again in two years. If you would like to help in planning a family gathering in the IL area, please email Crystal at [supermama95@yahoo.com](mailto:supermama95@yahoo.com).





# Research Committee Update

By: Meg Hefner

The Research Committee (RC) of the CHARGE Syndrome Foundation has been very busy recently.

- We approved funding of another Kim Blake study – this one surveying people about sleep issues and sleep apnea – information about participation can be found below this article.
- Melva Strang-Foster is heading up an effort to survey members of the RC to begin to prioritize research efforts, especially research efforts funded by the Foundation.
- Kristen Koehler is heading up the effort to work with an MIT (Massachusetts Institute of Technology) student, George Blake, who is designing a database so we can finally have a registry of people with CHARGE. The Foundation is funding the part of George's work-study that MIT does not cover.
- And along with that, I am working with Kevin Ballard in the Saint Louis University (SLU) IT department to put that database on a SLU computer. We are hoping these projects will make it possible to keep track of everybody out there with CHARGE. So every time someone is doing research, you don't have to fill out those first five pages confirming the diagnosis

Anyone who is interested in joining the RC, please contact [Meg@chargesyndrome.org](mailto:Meg@chargesyndrome.org). A reminder: all Foundation committee members must be current Foundation members. If you haven't renewed for 2010, you can do so by clicking on the Membership tab at the top of the Foundation homepage.

## Research Study:

### Understanding Sleep Apnea in Children with CHARGE Syndrome



Currently, there is very little information available concerning symptoms or treatment options of sleep apnea in the CHARGE Syndrome population. The principle investigator of this study, Dr. Kim Blake, is hoping to gather information in this area, as well as to determine the effects of tonsil and adenoid removal. Participants in this study will be asked to complete a short series of questionnaires regarding their child's sleep habits. The child must be between the ages of 0-14 with a confirmed clinical or genetic diagnosis of CHARGE Syndrome. If you are interested in participating in this study, please review the information sheet on the CHARGE Syndrome Foundation website at: <http://chargesyndrome.org/documents/BlakeApneaStudyInformation.pdf> and contact the research assistant, Carrie-Lee Trider at: [cr281800@dal.ca](mailto:cr281800@dal.ca) for more information.

## An Update on Gary James

By: Cheryl Rippa

Hello everyone! It has been 10+ years since I've written about my son, who was diagnosed with CHARGE syndrome at the age of 5 years. He is now 22 years old and on his way to becoming a fine young man. For an update: He had bilateral hearing loss, for which he wears hearing aids. He speaks well and does not use ASL. He also has microphthalmic coloboma of his right eye, with blindness of that eye only.



*Continued on next page...*

**Gary James**

*"The sky is  
the limit for  
you, my  
amazing  
son."*

*Continued from page 17*

His left eye has 20/25 vision (thank God). Gary also has OCD (obsessive compulsive disorder), which is somewhat controlled with the use of a prescription medication; mild retardation; and has reached his full growth height of 5' 1". He is a wonderful, Christian young man who is a very sociable, "people" person. He simply loves life and appreciates the simple things that most people take for granted.

As for his education, Gary graduated from our hometown high school last June, at age 21. He was mainstreamed for two classes, Science and Reading/Language Arts. He was in a self-contained homeroom to learn about such things as basic everyday living, art/woodcrafts, and basic computer and math skills. He also worked at Wal-Mart with his job coach and two other students two days per week. During his sophomore summer year, Gary successfully attended a Work Skills Prep program at The College of New Jersey for three weeks. He lived on campus in his own room and worked a few different job sites.

Now for the REALLY exciting part... Gary was enrolled at the Helen Keller National Center in Long Island, New York last August. He has since completed the 8-week evaluation and two 13-week sessions. His last and final session began on April 5th, and he will be finished in early July. I will try my best to express how much this wonderful opportunity has meant for him. He has grown in so many areas:

independence, perseverance, and emotionally. We (they) are now working with him to increase his stamina and independence by moving him into an on-campus apartment, called a SILP (supportive independent living program) in early May.

Gary works at the local food store in Long Island as a stock associate three days per week. Once he comes home in July, the store will refer him to our store here in New Jersey. He will have a vocational trainer with him in the beginning, then we are confident that he will be able to work independently at the store.

Gary still has much more to learn, of course, but he is determined, and we will help him get to where he wants to be in life. He is learning what it takes to succeed in so many areas, and he will do just that with the help of myself, my husband, and most of all, with the help of God. He knows that HIS Holy Spirit will always be there whenever needed for him for the rest of his life. Yes, having a child with CHARGE syndrome has always been a challenge, but I think we all can agree on one thing: the unconditional love and happiness that ANY person with special needs gives to all those who love him or her simply touches them in a very special way.

GO Gary James, the sky is the limit for you, my amazing son!

## Upcoming Regional Events

### Northeast Regional Seminar - Perspectives on CHARGE Syndrome:

**When:** September 24 - 25, 2010

**Where:** Quincy, Massachusetts

This seminar is sponsored by Perkins School for the Blind and is an in-service training for parents and professionals. Timothy Hartshorne, Nancy Hartshorne and Meg Hefner will be presenting. More information as well as registration dates will be coming soon—please continue to check the <http://www.chargesyndrome.org> website and ensure that the CHARGE Syndrome Foundation has your up-to-date email address so you will receive notifications about events such as these ones. Also, make sure [info@chargesyndrome.org](mailto:info@chargesyndrome.org) is listed in your address book so we don't end up in your junk email folder!



### Southeast Regional CHARGE/Deafblind Weekend

**When:** July 16-18, 2010

**Where:** MeadowView Conference Resort and Convention Center, Kingsport, Tennessee

For more information, or to register, contact: Jennifer Vick 615-936-2862  
[Jennifer.Vick@vanderbilt.edu](mailto:Jennifer.Vick@vanderbilt.edu)

MeadowView Conference  
Resort  
Photo Credit:  
Marriott.com



# Thank You

## THANK YOU FOUNDATION SUPPORTERS!

Click on the  
membership  
button below if  
you are reading  
online to go  
directly to our  
membership  
form.

Wells Fargo Community Support  
Campaign  
Microsoft Matching Gifts Program  
Abbott Laboratories  
Employee's Community Fund of the  
Boeing Company  
Network for Good  
Wanda Conness  
Ottawa Township H.S. Sign Language  
Club  
Michelle Langlors  
Yando's Store, Inc  
Deby Davis  
United Way of Kentucky  
Deby Davis  
United Way of Kentucky  
Mountain View Pediatrics, PLLC  
Community Health Charities  
Lacey Friedman  
Irene & Jim Lucas  
Bill Muir  
Diane Kelly  
Chari Roth  
Rolando Franco  
Jaime Bracken  
Jo-Ann Laframboise  
Geno Ray  
Joann Reynolds

Emily Murray  
Laurie Suter  
Michele Sinopoli  
Jackie and Mac Kenley  
Barbie Dull  
Christina Kis  
Ronda Lou McNeal  
Gary Nelson  
Wendy Keedy  
Ana Saruski  
Megue Nakazawa  
Ken Neuman  
Anara McCarthy  
Daniel Marena  
Charles Good  
Cathy Kauffold  
Nancy Crim  
Agustin Parodi  
**On the Special Occasion of  
Emma Bluestone's special birth-  
day**  
Sherie & Erwin Coyner  
**On the Special Occasion of Brad  
Andrew Miller's graduation**  
Steven & Barbara Lember  
**In Honor of Matthew & Brian  
Murray**  
Penn State NSSLHA

**In Honor of Erika Feather**  
Marc Sigismondo  
**In Honor of Christian Roberts**  
Bekkie Cobb  
**In Honor of Nicole Kowitt &  
Hannah Shikora**  
Roseanne Dyson  
Andrew Shikora  
**In Honor of Mallory Pigage**  
Ellen Elliott  
**In Honor of David Sirota**  
Bobbie Ringler  
**In Memory of Trey Chatham**  
Christine Hong  
**In Memory of Miles Bryce  
Sherman**  
Anne Kinsella  
**In Memory of Lucas**  
Sue Loyd  
**In Memory of Miles Sherman**  
Andrea Manchester  
Gerald Smith  
Carol Winkler  
**In Memory of Inez Mynatt**  
Lynn Morris

## HOW TO BECOME A MEMBER OF THE CHARGE SYNDROME FOUNDATION

Becoming a lifetime member or a yearly member has never been so easy. This year, we have introduced an online membership system that saves the hassle of printing and mailing forms to you and having you mail them back to us. We'll save some time and some trees in the process.

If you visit our membership page on the website at <http://www.chargesyndrome.org/membership.asp>, you will find all of the information and options about becoming a member. You will also see a button in the top right corner that looks identical to the one pictured at left. If you click on that, it will take you directly to the online membership form, where you can enter your information and either join the Foundation or renew your membership. It's very simple and quick! Also, if you are reading this online, you can click right on the button here in the newsletter to take you to the online membership form.

If you prefer to mail in your membership and payment, that option is also still available. There is a printable form on the membership page that you can print off, fill in and mail back with your payment.

We appreciate your support and look forward to having you as members!

**MEMBERSHIP**

Please Click on the Button Above  
to Join or Renew

# Thank You

## Second Annual CHARGE It for CHARGE Donors

### In Memory of William "Billy" Taylor

Pam Ryan, DeafBlind Program,  
Hilton Building Perkins

School for the Blind

CHARGE Syndrome Foundation  
Exec. Committee

Daniel Stusser

Lori Swanson

Susan & Joel Wolfe

### In Memory of our daughter, Rileigh and Adrienne Antaya. Always in our hearts

Cynthia Antaya

### In Memory of Barbara Drinko

Thomas R Drinko

### In Memory of Colin Luke Smith

Laura Kerekes

Robyn Semliatschenko

Jeanne, Gary and Liam Smith

Nicole Vinciguerra

### In Memory of Bridget Vannucci

Michael Vannucci

### In Memory of Joshua Roy Kurby

Rosalie Kurby

### In Memory of our precious grandson, Colin Love Always!!

Thomas Mills

### In Memory of Trey Chatham

Holly Roberson

### In Honor of William E Taylor, Sr

Houston West Chamber of  
Commerce

Terri Bieber-Christen Stilwell of  
ARTreach

### In Honor of Finley Roth

Robin Bodek Rosenbaum

Mary Briggs

Erin Ford

Valorie Hobson

Robin O'Meara

### In Honor of Doug Roth

Lloyd & Betsy Bell

Bruce Combs, M&M BS

Michael Coyne

Lynn Koster

Jeffrey McCormick

Kevin McGreal

Helen Mooradian

Jennifer Myers

### In Honor of Colin Smith

Denise Gillman

### In Honor of Aaron Hiscutt

David Goldstein

Kim & Carson Lane

### In Honor of Trey Chatham

Patrick & Donna Downey

### In Honor of John Thomas Chatham – son of Sharon & John Chatham, Marietta, GA 3/23/09-4/4/09

Karen Lee

### In Honor of Douglas Charles Roth

Marcia Roth

### In Honor of Eli

Mary Strandberg

### In Honor of Halyn Jones, our sweet CHARGER

Melva Strang-Foster

### In Honor of the Hiscutt Family

Kimberly Wellman

### In Honor of Justin Murray

Alice Russo

Paul & Pamela Dowd

### In Honor of David Sirota

Valery & Sofia Bazarov, &  
Lyubov Eydman

Alex Ranger and Family

Ruth Ringler

Julie Steinberg, UBS

### In Honor of David Sirota. I am so proud of the strides you have made and thankful to be a part of your life.

Melanie Vega

### In Honor of David Sirota love the Wirzman Family

Jerald & Kim Wirzman

### In Honor of the Sirota Family

Alyssa Weiss

### In Honor of our great nephews Matthew & Brian Murray

Leba and Neal Yolin

### In Honor of Gracie's 7<sup>th</sup> Birthday

Helen & Steve Colburn

### In Honor of Gracie Swann

Kimberly Diamond

Jacqueline Dominick

Cameron Higley

Sheila Lopez

### In Honor of Grace Dagley

Elise McCaffrey, Hilton Head  
Island Early Childhood Center

### In Honor of James A and Catherine M Roth

Mark Drinko

### In Honor of your family by Robert & Lori Gelman

Robert Gelman

### On the Special Occasion of Gracie's 7<sup>th</sup> Birthday!

Jacqueline Dominick

### On the Special Occasion of Trey's Birthday

Lisa Fore

### On the Special Occasion of Miss Gracie Swann!!!!

Cathy White

### On the Special Occasion of your 7<sup>th</sup> birthday, Gracie. Your Mom is right – you are a teacher, and I'm glad to have been one of your students. Many, many more happy birthdays are wished for you, sugar girl. Lots of love. Judy

Judy Rainbow

### On the Special Occasion of Lori's 40<sup>th</sup> Birthday!

Georgette Morales

### To Sirota's Family From Meg Hawkins

Lisa Hawkins

### To David and his beautiful family....From your preschool buddy Alexis and her family

Catherine Rose

### We love our Douglas

Jeanie M Roth



# Upcoming Newsletter Topics

## Survival Strategies: CHARGE Style

Recently, the topic of 'survival strategies' was brought to our attention as a possible topic for a newsletter.

What types of organizational strategies do you use for tracking you or your child's health information? Appointments? Family schedules? General household/cooking/cleaning tips? Do you use lists? Spreadsheets? Do you work outside the home? How did you organize things for childcare? We want to know all of the great tips and tricks of how you & your family manage!

## Topics of Interest to our Readers

We would love ideas from our readers about what issues or topics they would like explored in future newsletters.

### CHARGE In the News



Have you seen a great article about someone with CHARGE or about CHARGE syndrome itself in the news (either online or in a newspaper/magazine)? Send it along!

### Celebrating Accomplishments

Do you have a story about yourself or your child that you would like to share? A story they wrote or a picture they drew? Send it to me with a picture if possible so we can share in you or your child's accomplishments.



Please send any stories and pictures to [lisa@chargesyndrome.org](mailto:lisa@chargesyndrome.org).



## 10th International CHARGE Syndrome Conference

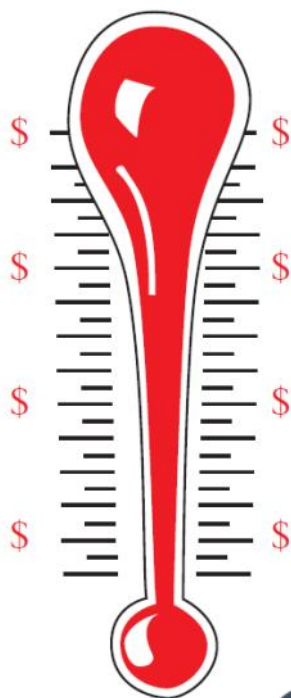
Next year's conference will be held on July 28-31, 2011 in Orlando, Florida at the [Rosen Shingle Creek Hotel](http://www.rosenshinglecreek.com/rosenShingleCreek.asp) (<http://www.rosenshinglecreek.com/rosenShingleCreek.asp>)

Save the date and start making plans to attend what promises to be another outstanding conference! A link will be available on the [conference information page](http://www.chargesyndrome.org/conference-2011.asp) <http://www.chargesyndrome.org/conference-2011.asp> in early 2011 to register and make your hotel reservation.

Questions? Contact [Janet Murray](mailto:janet@chargesyndrome.org) ([janet@chargesyndrome.org](mailto:janet@chargesyndrome.org)) - 2011 conference chair



# HOW HIGH CAN WE GO?



2010 may be the year of the CHARGE events. We are off to a fantastic start with runs, a skating party, a golf outing, bean bag toss and even a lemonade stand just to name a few! Meanwhile, our Second Annual Charge It for CHARGE campaign has raised over \$71,000. We are on our way to passing 2009!

Thanks to everyone who has hosted an event and participated in our fund-raising activities. It is never too late to plan an event that interests you. Please keep the emails and ideas coming. Contact our fundraising chair, David Wolfe at [david@chargesyndrome.org](mailto:david@chargesyndrome.org) with any ideas or questions. Help us make 2010 our best year yet.

## Come Join Us!

### **CHARGE Syndrome Foundation, Inc.**

141 Middle Neck Road  
Sands Point, NY 11050  
USA

Phone: 516-684-4720  
Toll Free: 800-442-7604  
Fax: 516-883-9060

#### Annual Membership

Family \$25  
Professionals \$40

#### Lifetime Membership

Silver \$250  
Gold \$1,000  
Platinum \$5,000  
Diamond \$10,000



Email: [info@chargesyndrome.org](mailto:info@chargesyndrome.org)

Visit us on the web: [www.chargesyndrome.org](http://www.chargesyndrome.org)