The Last Piece

When I was invited to speak with you today, I was asked to tell about my journey of learning about having CHARGE Syndrome only after the birth of my son who is also diagnosed with CHARGE Syndrome. I was 27 years old. 27 years is definitely a long time to go without knowing that missing piece to a very complex puzzle I had lived my entire life. However, it is my hope that you will learn just how it has affected my life.

I am an adult with CHARGE Syndrome. I was born with bilateral choanal atresia which was corrected after birth. I was also born with bilateral moderate-severe hearing loss which has intensified to a bilateral severe-profound loss over years. I grew up oral and learned how to sign in college. In addition, I was born with bilateral blocked tear ducts, which was corrected through multiple surgeries. I endured six years of growth hormone therapy and I have hypothyroidism. The anterior part of my pituitary gland is missing. I have hydronephrosis of both kidneys which was only discovered four years ago. My left kidney is significantly smaller than my right and is suspected has been that way since birth. In the last nearly 30 years, I have endured countless surgeries, multiple medical treatments, and many more tests.

I enjoy listening to music, hiking, biking, and being with my family. I enjoy following Boston Red Sox baseball and other Boston sports. I love Italian food. I'm a true Bostonian living in the south so I am always the person with a funny accent because for some reason, I'm always the one who can mix the word "cah" in the same sentence with "ya'll". I love going to the beach, watching the waves, and building sand castles. I am an adult with CHARGE Syndrome.

Before July 13, 2007, I had always felt there was a missing piece to my life. This especially occurred after I moved from the Boston area, where I was born and raised, to Washington DC. I had to find new specialists to follow me on a variety of medical conditions and I had to answer many questions. Nine times out of ten, the new doctors asked, "is there a name for all of this?" My answer, "no".

July 13, 2007 came a day full of excitements and emotions for my husband, Keith, and I. It followed two medically complicated pregnancies. The first resulted in the loss of our identical twin daughters, Rileigh and Adrienne to premature birth at 20 weeks gestation. Our pregnancy with Brady was doubly complicated with 4 months of strict bed rest, insulin dependent gestational diabetes, concerns with my kidneys, and monitoring of his brain as the ultrasounds were showing a cyst. When Brady came six weeks early, and was screaming when he came out, we both let out a huge sigh of relief. The gift we had worked so hard for was finally here!

Once Brady was settled in the Neonatal Intensive Care Unit, Keith got the surprise of a life time. Or rather, I should say, the NICU doctors got a surprise of a life time! They told Keith about Brady's breathing issues and very gently told him that Brady had a severe respiratory condition called bilateral choanal atresia. Now, I don't know what the doctors were expecting to hear but I'm guessing it wasn't "huh. My wife has that too!" I can only imagine the looks on their faces and the shock running through their minds as it is not widely known for many of the conditions of CHARGE Syndrome to be passed on within generations of one family.

So, at 27 years old, I was bombarded with questions from geneticists about my entire life. Every surgery, growth hormone treatments, and every medical test was examined. I was thankful that in just the day after Brady was born, my parents arrived in Washington, DC to be with us. I don't think I could have answered half of the questions I was asked without their presence. I have to laugh sometimes when it is assumed that I know exactly what it was like to be born with Choanal Atresia so I can highly relate to Brady. The bottom line was, everyone wanted to know why I had not been diagnosed. All I knew was that I had not, but with Brady's birth into this world, I finally felt like the last piece to this big panoramic puzzle was put into place. CHARGE Syndrome.

Growing up, I feel that I was given every opportunity to grow and develop as was expected of me. My parents never once allowed my challenges to set me back. I was a competitive figure skater for 12 years, winning multiple awards. I played little league baseball in middle school. I enjoyed track and field in high school. When times were difficult, especially in school, my father always had the final say; "all you can do is your best". Throughout my life, this phrase never seemed to leave my mind. I have carried this with me and it truly has helped me get through some difficult times in my life. School was not always an easy journey for me. I had good years of school, earning honor roll grades and felt quite successful. Other years, I struggled. High school started out as a challenge as I chose to attend a private school. I strongly feel that part of the difficulty at this private school is that the teachers did not take a moment to truly understand my disability. Perhaps now we know there was much more for them to understand than simply hearing loss. I transferred to the local public school for the start of my junior year of high school where I excelled in every subject and earned multiple scholarships for college. I continued to grow through college and pushed myself with the "all you can do is your best" attitude. In 2001, I graduated from college with honors and accepted an academic scholarship for graduate studies in the field of School Psychology at Gallaudet University in Washington, DC. Today, I hold a Virginia state license in school psychology and I am a nationally certified school psychologist. I have been employed by Fairfax County Public Schools in Northern Virginia for the last five years.

I truly hope that I can pass on the simple line of advice that my father gave me, to Brady as he grows. The support I have received from my parents and older sister growing up far exceeds what would have been expected of them. They taught me how to advocate for myself and to be proud of who I am. I have been able to embrace my disability for as long as I can remember and I greatly attribute this quality to my family. They never once told me I could not do something because of some medical issue or because I could not hear entirely.

I have to say that I have learned a lot about my condition with CHARGE Syndrome. However, I was not always accepting of this diagnosis. When Brady was first born, it was extremely difficult for me to process the fact that, first, I even had this syndrome, and second, that I had passed it on to my son. When I was first informed, all that I could think about was all of the surgeries, medical tests, and countless hospital visits I had endured as a child and teenager. I definitely did not want Brady to have the same life as I did but here we were, with him having already so many of the same medical conditions I was born with. The prospect of him enduring many of the same things I went through was painful to me. Things ran through my mind but one of the main issues I knew I would have to face is within all of this medical care he would require, how was I going to explain to Brady when he was older that he has CHARGE Syndrome because of me?

Something people know little about is that the day my parents left from visiting us after Brady was born, just a few days after his birth, Keith and I sat in my hospital room for a number of hours discussing all of this. I held a tremendous amount of guilt. However, as we talked about things, Keith convinced me that we had to take this one step at a time. First off, we were truly overjoyed that our son was here on this earth with us. Second, we will have to learn about all of these medical conditions and treatments together. We did just this and it has certainly gotten us through so much over the last two years with Brady. Keith told me "when the time comes to explain to Brady about what CHARGE Syndrome truly is, we'll do it together". I know in my heart it will not be easy, but I hope that in the long run that when Brady knows he has this complex medical disorder, it will impact his adult life differently than it did mine.

Keith and I spent 56 days in the Neonatal Intensive Care Unit watching Brady go through 2 major surgeries, many issues with breathing, a severe bowel infection which we were told he had only a 30 percent survival rate, sodium deficiencies, and most of all, getting ready for a world of CHARGE Syndrome outside the hospital. It often strikes a person as odd when I explain that even I had to get ready for this. Mentally, I had

to gather the strength and knowledge on how to care for my son. I felt like I was catching up on 27 years worth of knowledge, finally making connections, and being able to learn about my son all at the same time.

Through the last two years, we've learned patience is definitely a virtue. With all of the advice we have received, we've had to sort through and pick out what was necessary for Brady. We have reminded a lot of people that Brady is not exactly like me. Through Brady, I have learned that no child with CHARGE is exactly like another. The perspective I take is that you cannot compare one child with CHARGE Syndrome to another because likely, one child has one slightly different condition than the other. While Brady and I are alike, we are quite different from each other in the characteristics of CHARGE that we exhibit. Brady has severe vestibular dysfunction, low set ears, low muscle tone, requires orthotics for support with walking, reflux, and he is a high risk of aspiration pneumonia. Brady learned to walk before I did at 19 months. I did not do so until after 24 months of age. I am still amazed that with his vestibular dysfunction, he is able to walk as well as he can today. Brady has learned much more sign language than I ever did as a child. However, we still continue to struggle with the comparisons of how much Brady is like me. We've chosen to focus on not fighting back and listing all the differences, but to channel our energy at helping Brady to grow and become the individual he will become.

In the past two years, I have been asked multiple times, if I knew I had CHARGE Syndrome, would we have still went on to conceive Brady? Truthfully, it had taken 27 years to get a diagnosis. If Keith and I did not have Brady today, I think I would have lived another 3 years without a diagnosis. I don't like to look back on the "what ifs" about our family situation. Rather, I focus on all of the wonderful people who have come into our lives as a result of Brady's birth. We've met doctors, nurses, physical therapists, occupational therapists, speech and language therapists, ASL specialists, and orthotics specialists who have helped us guide Brady on the path he is set on today. In addition, we've made a new network of friends through the CHARGE Syndrome listserve. We definitely do not feel like we are alone in this little known world of CHARGE Syndrome.

We were even fortunate to meet another family through Brady's early intervention program he attended from the spring of 2008 through the spring of 2009. We met the Gerstein family last summer and truly, that was day we finally felt we made a connection with another family. Brady was in an early intervention program for children who were deaf and hard of hearing. Having a child with multiple disabilities in a room full of children who were "typically developing" was not easy. When we met the Gerstein family, we were not overjoyed at the fact there was another child with this rare syndrome, but overjoyed that we finally had friends we could talk to who knew exactly what we were talking about. Thus began a relationship that will likely last a lifetime. Tomer and Brady both have CHARGE Syndrome but are quite different from each other. I believe we have all enjoyed learning from each other, offering advice when one asks for it, and supporting each other as family units through the good times and the bad.

I also feel fortunate that we have found one physician for Brady who has vested time and interest in learning about CHARGE Syndrome. Dr. Rachel St. John is the director and pediatrician for the Kids Clinic for the Deaf at Georgetown University Hospital in Washington, DC. When we first brought Brady to her, we were in the process of changing pediatrians due to a bad experience with the first one we had picked out for Brady. While Dr. St. John expressed she had heard of CHARGE Syndrome, she did not know much about it. However, when we returned to her office a week later, she had researched, made phone calls, and read so much information about CHARGE Syndrome. As Brady's parents, we are comfortable with calling or emailing her with questions about anything related to Brady's general health or about CHARGE Syndrome. Dr. St. John has by far surpassed our expectations as a physician and is truly a model for any doctor who treats any child with a rare genetic syndrome.

Often, when I am talking about CHARGE Syndrome and my connections to it, I am asked how this diagnosis has affected my marriage and my relationship with my husband. I truly feel that I am blessed to

have found the perfect person to spend the rest of my life with. We recently celebrated our 6 year wedding anniversary, though we have spent the last 14 years together. Through our journey together, Keith has taken the time to get to know me and understands all of my medical conditions as I do. I often joke with him that on our wedding day, June 28, 2003, that Keith thought he knew everything there was to know about me. However, on July 13, 2007, we got the surprise of a life time. There was more to me than anyone knew! He always ensures me though that he was in this relationship and family to ensure that we are happy and for the most part, healthy. Keith is my rock. I know that when things become too emotionally overwhelming for me, he steps up and puts things in order. He has helped me get through the worst days of my life and has celebrated the best days with me. When we lost our daughters, Rileigh and Adrienne, to premature birth on March 8, 2006, we grew closer together and had a newfound love for each other that neither one of us knew existed before then. When Brady was born, we found strength that we didn't know we had. This strength helped us grow as a family and has given us three a bond that will last a lifetime.

Some people ask how I identify myself. In many ways, I still feel like I am torn between two different worlds. For one, I identify myself as a parent for a child with CHARGE Syndrome. I, like so many of the parents in this audience today, learn something new about this syndrome everyday and find new things out about my child, medically or educationally. I post questions and concerns on the online listserve which is definitely a huge part of the support I seek. I want to know all the information I can get so that I can help Brady in the best possible way.

On the other hand, here I am, an adult with CHARGE Syndrome. There is so much I still have to learn about myself while I am learning and growing with Brady. I have lived nearly my entire life with this rare syndrome and did not even know it. I am fascinated by the in's and outs of CHARGE Syndrome and learn something new every day; not only to benefit myself, but to benefit Brady. As an educator and parent, I vest my time in learning about CHARGE as much as I can. I want to make sure that I am not empty handed for doctors I see now and doctors I will see in the future. In this aspect, I'm glad to finally have a diagnosis and something to call "all of this".

I find myself fortunate that I work in the education world as a school psychologist. CHARGE Syndrome is definitely not known in this realm. I feel like if I can start educating just the people I work with in Fairfax County Public Schools in Northern Virginia, I am starting out on the right path of bringing more awareness to CHARGE Syndrome. I have done just this, using my strength and knowledge to educate those around me. Many of my co-workers follow our family blog and are continuously asking questions about CHARGE, which brings a smile to my face. If I can educate just one more person about CHARGE Syndrome, that person can educate another. The more education that takes place, the better off every child with CHARGE Syndrome will be and the more options they will have educationally which to me is equally important as the medical options that every parent searches for.

Part of my personality involves giving back. For example, each year, our family participates in the March of Dimes March for Babies as a way to give back to all that the March of Dimes has done to help us honor the memory our daughters and to celebrate Brady's life. When I found out about my diagnosis of CHARGE Syndrome, after I had time to process it and accept it, I decided I needed to somehow begin to give knowledge to others about it. I started out by learning as much as I could about this rare genetic disorder and I continue to do so much every single day. However, more recently, I felt that my nomination to the board of directors of the CHARGE Syndrome Foundation would be the perfect way to expand my giving back. I am happy and excited to be more involved in the Foundation that has provided so many opportunities to all of the children and young adults diagnosed with CHARGE. Since I am able, I want to make contributions to the goals of this Foundation. I want to be a voice for all of those who have CHARGE Syndrome.

I believe the bottom line for all of us parents is the love we provide for our children. I see the love that Keith and I give Brady every single day and how it helps him grow. He has come such a long way and I am positive that all of you can say the same for your children. While there are such differences in all of our children, we do live with one commonality. CHARGE Syndrome. We can learn from each other and teach each other about our children. The next step is to teach the world about CHARGE Syndrome, what it entails, and just how unique our children truly are.

Thank you.