



# Welcome!

Dear Parents,

Congratulations on the birth of your baby!! We are looking forward to meeting your new addition.

This is a time to celebrate the new addition to your family! This new life will bring joy to your family that you did not know was possible. Your baby is a baby first, with his or her unique personality and talents. Your baby will reach each new milestone when they are ready. The hurdles may take much longer to overcome but children with CHARGE will work harder than most and bring such excitement as you watch them succeed. Never underestimate your child's abilities.

The CHARGE Syndrome Foundation is an organization of parents and professionals who are available to support you as you enter into this new and challenging phase of life, parenting or caring for a child with CHARGE syndrome.

Our Director of Outreach, Jamie Goodpaster, is available to provide support, information, connections, and resources. You can reach Jamie at [jamie@chargesyndrome.org](mailto:jamie@chargesyndrome.org) or 855-5CHARGE (1-855-524-2743).

We invite you to join the Foundation and receive our mailings. We can put you in touch with other parents who have been through what you are going through. They can guide you on your journey. We encourage you to refer to the website often for information.

In the meantime, enjoy your child and remember to take care of yourself. Please remember that this child needs what every other child needs – to be loved unconditionally, accepted, and valued just for being them.

We look forward to hearing from you soon!!

The CHARGE Syndrome Foundation



## Welcome from the Director of Outreach

Dear Parents, Guardians and Caregivers,

Welcome! Whether you are a new parent or new to our Foundation, this is a packet that will help guide you on your journey. Please do not use this packet in isolation. I encourage you to reach out and connect to the CHARGE Syndrome Foundation, parents, professionals and people with CHARGE syndrome. As Director of Outreach, my job is to help you make those connections, access resources to guide and be a source of support.

CHARGE syndrome is complex and requires parents to be knowledgeable in many areas. To obtain this knowledge you will need support and access to good information. Well-informed parents ensure success for their child. Large or small, success can be measured in many ways. Your child deserves the best so they can achieve milestones. A child with CHARGE syndrome faces many challenges, but their potential to thrive and achieve should never be underestimated.

This is your Foundation and I am here to support all families that have a loved one with CHARGE syndrome. You can reach me at my toll-free number 1-855-5CHARGE (1-855-524-2743) or at [jamie@chargesyndrome.org](mailto:jamie@chargesyndrome.org). Please make sure you visit our web site at [www.chargesyndrome.org](http://www.chargesyndrome.org) and view the resources we offer to families. I encourage you to join our Foundation to support our mission of providing support and information while promoting awareness and research. I look forward to speaking with and connecting with you!

Warmly,

Jamie Goodpaster  
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## Who Are We?

Dear Parents, Guardians and Caregivers,

The CHARGE Syndrome Foundation, Inc. is an organization of parents and professionals that grew out of a desire to help make life a little easier for families living with CHARGE syndrome. The Foundation is a non-profit organization that provides information to families and professionals working with individuals with CHARGE. We also sponsor a biennial conference and fund CHARGE syndrome research. To offset some of these expenses, our annual membership fee is \$30. Lifetime memberships are also available. We invite you to [become a member](#) of the Foundation.

You can find [additional information](#) about the Foundation on our website. You can contact us by calling (800) 442-7604 or by email [info@chargesyndrome.org](mailto:info@chargesyndrome.org).

The services provided by the Foundation include:

**The CHARGE Syndrome Foundation Website**, [www.chargesyndrome.org](http://www.chargesyndrome.org), provides information on CHARGE syndrome, the Foundation Board of Directors, committees, conferences, and resources. It also provides links to sites for support such as a families page, deaf-blind resources, and links to our Facebook page and Facebook group. You can also read old issues of our newsletter, [CHARGE Accounts](#).

**The CHARGE Syndrome Management Manual for Parents** is a must for every family. The manual includes medical information for both families and professionals, developmental and educational information, a glossary, and a listing of resources. The Manual is available for download in [English and Spanish](#).

**Parent Support:** Jamie Goodpaster, our Director of Outreach, is available to provide support, information, and resources. Jamie can also put you in touch with other families in your area. You can reach Jamie at 1-855-5CHARGE (1-855-524-2743) or by email: [jamie@chargesyndrome.org](mailto:jamie@chargesyndrome.org).

**Biennial Conference:** The International CHARGE Syndrome Conference is held every two years. The purpose of the conference is to bring together families and professionals who have a common interest in CHARGE syndrome in order to discuss the latest medical, educational, and advocacy information available and to share experiences for mutual support. There are scholarships available to assist those families attending their first conference. Please check the [Conference Information Page](#) for information about prior and upcoming conferences.



# CHARGE Syndrome Fact Sheet

CHARGE syndrome refers to a specific set of birth defects, medical problems, and developmental issues. The most distinctive birth defects are coloboma, choanal atresia and characteristic ears (external ears and small/absent semicircular canals).

- **Diagnosis** should be made by a medical geneticist. Diagnosis is based on key features, ideally with DNA testing for *CHD7* mutations. Key features:
  - Coloboma
  - Cranial nerve abnormalities
  - Choanal atresia
  - Heart defects
  - Characteristic external ears
  - Esophageal defects
  - Small/absent semicircular canals
  - Genitourinary abnormalities
  - *CHD7* gene mutations
- **Incidence:** One in every 8,000-10,000 births. Every person with CHARGE has a unique set of features. There is wide variation in physical features and cognitive ability.
- **Cause:** Mutations in the *CHD7* gene on chromosome 8 are found in 80-90% of cases. There is no relationship to sex, race, nationality, religion, socio-economic status, or prenatal exposure.
- **Recurrence:** It does not usually run in families. Recurrence risk to unaffected parents is 1-2%. If a parent has CHARGE Syndrome, the risk to a baby is 50/50.
- **Sensory deficits:** Most individuals with CHARGE have difficulty with hearing, vision and balance. This results in delayed motor development and communication. The educational term for combined vision and hearing deficits is “deafblind.”
- **Cognitive ability & testing:** Many have decreased cognitive abilities, but 30-50% have normal intelligence. Intelligence of children with CHARGE is often underestimated due to the effects of combined hearing, vision and balance issues. Testing, therapies and educational intervention **MUST** take into account hearing, vision and balance status.
- **Lifespan:** There is an increased mortality, especially in the first two years. Although individuals with CHARGE remain medically fragile, lifespan can be normal.
- **Outcome:** Individuals with CHARGE need medical care appropriate to their particular features. In addition, early intervention and appropriate and challenging educational and vocational programs specific to their sensory needs are imperative. Although there are many problems, children with CHARGE can survive and become healthy, happy citizens.



# Frequently Asked Questions

## New diagnosis

You have been told that your child has or may have a condition called CHARGE syndrome. Your baby probably has multiple medical issues and may still be in the hospital. This is a scary time for you and your family. All parents have questions about the diagnosis and what it means for their child and their family.

We wish we could tell you it will all go away. Unfortunately it won't. You may have some difficult days ahead of you. But please remember, there are many of us who have been there, done that. Please do not hesitate to come to the CHARGE Syndrome Foundation for advice and support.

## Why do they think my child has CHARGE?

Your child likely has several birth defects and may already have had some genetic testing, such as chromosomes or a microarray. If those tests have not provided the answer and the baby has coloboma, choanal atresia, or distinctive ear findings, CHARGE syndrome should be considered. Most babies with CHARGE also have other problems (heart, trachea, esophagus, etc.). Your medical geneticist should be able to tell you why the diagnosis of CHARGE is being considered.

## What is CHARGE syndrome?

CHARGE is a recognizable genetic syndrome most often caused by mutations in the *CHD7* gene. It occurs in about 1 in every 10,000 births. Most individuals with CHARGE have distinctive features, including coloboma, choanal atresia, and/or ear abnormalities, along with other birth defects. More information on features [here](#).

## Why is a diagnosis important?

A diagnosis provides an explanation for why your child has multiple issues. It tells the doctors what other potential problems to look for in your baby and gives you information about what caused your baby's problems and whether it could happen again.

## How do they make a diagnosis?

Confirming a diagnosis of CHARGE requires exams by various specialists (genetics, ophthalmology, cardiology, ENT, audiology), imaging (kidney ultrasound, brain & inner ear MRI) and tests (*CHD7* DNA testing). A medical geneticist should put together all of the information to determine if CHARGE is the best diagnosis for your child.

## Is there a test to confirm CHARGE?

Yes and no. CHARGE is still diagnosed by a medical geneticist primarily based on clinical features. *CHD7* gene testing is very helpful and should be ordered if CHARGE is being considered. This test takes several weeks to be completed and does not always provide answers. If a mutation in *CHD7* is identified in a child with CHARGE features, the diagnosis is confirmed. But negative *CHD7* testing does not rule out CHARGE. A significant number (~20%) of individuals with CHARGE do not have identifiable mutations in *CHD7*. Testing may become better in the future. For additional information, see the [CHD7 FAQs](#).

## Why are so many different specialists seeing my child? Who is in charge?

Most children with CHARGE have complex medical issues. Many different specialists will be following your child. Communication between all these specialists and with the parents is often less than ideal. If you can identify one specialist who is in charge of coordinating your child's overall care, that may help. It may be a cardiologist, a geneticist, or someone else. Some hospitals have care coordinators to help. Sometimes it is possible to have a "team meeting" to bring together all of the specialists to explain (to you and one another) the plan of care for your child. The Management Manual for Parents can help you understand the role of each specialist.

Don't forget your pediatrician. Children with CHARGE are children first. A pediatrician is important for all of the regular things like immunizations, weight checks, ear infections and so on. Your pediatrician may also be an advocate for you and your child in the complex medical system.

## How does CHARGE syndrome happen?

Most cases of CHARGE are caused by a mutation, or change, in the gene *CHD7*. This mutation usually happens for the first time in the person with CHARGE – it is usually not inherited from either parent. Not all people with CHARGE have a change in the *CHD7* gene – other genes for CHARGE may be discovered in the future. CHARGE is NOT caused by any known exposures during pregnancy nor is it related to sex, race, nationality, religion, or socio-economic status.

## Will it happen again?

Probably not. Most of the time, the *CHD7* mutation happened only in the sperm or egg that formed your child with CHARGE. In rare cases, the mutation happened in the gonads – the organs that make sperm or eggs. In those rare cases of gonadal mosaicism, there is a recurrence risk. Overall, if we look at all couples who have one child with CHARGE and look at what happens in the next pregnancy, 98% of the time, the next baby is fine. About 1-2% of the time (1/50-1/100), there is another baby with the same *CHD7* mutation as the first child with CHARGE.

## What about my child's children?

A person with CHARGE has one gene for CHARGE and one normal gene. The chance of passing on the CHARGE gene is 50-50 for an individual with CHARGE. People in the family with the same *CHD7* mutation may or may not have similar features.

## Can it be diagnosed before birth?

If a *CHD7* mutation is identified in a person with CHARGE, it is possible to test other people (or pregnancies or pre-implantation embryos) for that same gene. One reason to do *CHD7* testing in a child with CHARGE is to make it possible to look for it in other family members.

## Will my child see and hear?

Most children with CHARGE have limited vision and/or hearing. Many parents are told their child will be “blind” or “deaf.” Legal blindness does not mean the inability to see anything. Even significant hearing loss can often be helped with aids of various sorts. In the early stages of a newborn’s life, it is difficult to predict eventual vision and hearing abilities. The early predictions you are given may not turn out to be accurate. Routine visits to pediatric ophthalmology, ENT and audiology will help uncover your child’s abilities.

You as parents or caregivers know your child best. Doctors see your child for short periods outside of the home. Keep doctors and therapists informed of progress you see at home. Take comfort in knowing that these kids learn to naturally compensate by using whatever vision or hearing they have. For them it is not a loss – it is all they know.

## How does CHARGE affect cognitive abilities?

The sensory losses (hearing, vision, balance), time lost to surgeries, and frequent illness have a huge effect on the child’s exposure to the stimulation that shapes cognitive abilities and other skills. We expect children with complex medical issues to be delayed. But catch-up often happens.

Because of the sensory deficits, especially vision and hearing, communication is a big concern. A communication system must be established before cognitive ability can be determined. Intelligence is routinely underestimated due to vision, hearing, learning, motor and/or speech disabilities. Take advantage of all services available to help your child reach full potential, whatever that may be. See the Developmental sections of the Management Manual for information on [sensory deficits](#), [assessment](#), [education teams](#), and [therapies](#).

## What does the future look like for my child?

Although children with CHARGE have many challenges, they can survive and become healthy, happy citizens. Doctor visits and medical problems taper off and/or change as your child grows. Accept that you can’t predict what will happen and enjoy today while doing what you can to prepare for the future. NEVER underestimate your child’s abilities. Be involved, interact, and enjoy. As hard as it may be at times, they grow up fast, overcome many obstacles, and will make you proud.

## What services are available?

Most states have Early Intervention (EI) programs for children from birth to three and school-based programs starting at age three. Most EI programs include services such as physical therapy (gross motor skills such as crawling and walking), occupational therapy (fine motor skills such as pinching and grasping), speech therapy, education, and possibly vision and hearing services.

In the US, every state has a [DeafBlind program](#). Your child does not have to be deaf or blind to qualify. Most children with some vision loss and some hearing loss qualify for services through the DeafBlind program starting in the newborn period.

If you have not heard of these programs yet, ask to speak with a hospital social worker and ask her to help you find out more about what is available in your area.



## Parent Stories

### **Andrew's Arrival** by Minnie Lee Lambert

With my Momma and Doug by my side, I was in a bed at East Copper Medical Center awaiting the arrival of my little Clemson Tiger. I had no clue that our lives would be turned upside down. I had no clue that anything was wrong with Andrew.

They had given me medication to induce my labor last night and broke my water around 11 am this morning. Today was a slow day with very little progress. Andrew was trying to come out looking up and was not turning. This epidural was Hell.. Took that guy over 14 tries to get it in my back and it felt like fire was shooting out of my feet. I tried not to break Doug's hand but this was hard. Momma stood behind the bed and rubbed my head and told me I was doing so good. The talks of a C-section have started. The thought of surgery scares me but there really is no turning back now. Doc says we will wait a little while longer but by 6 pm if he is not here, we are going to the OR.

Doug is dressing in a white suit and looks like he is scared to death. They are getting ready to wheel me out. I have never been in the hospital before and now I am heading into surgery! Why couldn't this little guy just turn and come on into the world?! Why couldn't my body have just done what it should and he would be here by now?!

**6:48 pm.** Andrew makes his entrance. The doctor told me that he needed to tell me something about him. I looked up at him and laughed and said "What, He's a Gamecock?" I could tell by the look on his face this was not the Clemson/Carolina jokes we had been having all day. Dr. Osbourne tells me that my baby has a cleft lip but not to worry because he has a good friend that will take very good care of Andrew. Doug is crying. I am crying. Is this a dream? How can this be? They bring him around and I see his face. He is mine and he is BEAUTIFUL! <sup>[1]</sup> Then they take him away. After surgery I see Babby. She is crying. I ask about his lip. No, something else is wrong she tells me. His feet were BLUE. Blue like blueberries. This has to be a dream. No way this is real. Where is my baby? I need to see him NOW!

They bring him to me and I'm loving him through the tears. Momma is crying and Doug is being very quiet. The nurse said she will take him back to the nursery soon. They have to put a feeding tube in his nose because no one is here to help me learn to feed him. We were not expecting this!

They took him to the nursery and they never brought him back. By now I come to realize that something else must be wrong. The attending physician came in and sat on the side of my bed. A doctor sitting on the bed is never a good thing. She told me that they thought he had a heart murmur but when they went to put the feeding tube in his nose his O2 stats plummeted. The helicopter is coming. He is going to MUSC. I am terrified. I want my baby!



**10:20 pm.** The door opens. Two nurses, three flight crew members and the attending physician come into my room. They push my bed into the corner and bring in the long transporter that looked like something from Star Wars. It had O2 tanks on the bottom and small arm holes on the sides. Lying inside it was the little 7 pound, 13 ounce baby that had just broken my heart into a million pieces. He seemed a mile away. I could not even reach him so I rubbed the little blanket that he was wrapped in. They gave me a picture of him then they took him away. My heart left with him. How can they take my baby and give me a picture? I don't want a PICTURE! I got upset to hear the other babies in the hospital crying. I yelled at Doug and told him to shut the door! I did not want to hear someone else's baby I wanted MINE!

**Today.** Andrew is my little miracle. MY HERO! I so openly share our lives and his story because I want everyone to know how truly special he is. If he can come through all of this, there is NO LIMIT to what he can do. His life has been a battle since day one, but anything worth fighting for is definitely worth having!

I am thankful for each and every day and all of the pebbles along the way! I am thankful that the Lord chose me to be Andrew's momma!

## A Well-Deserved Snow Day by Lisa Weir

How fortuitous to wake up this morning, the morning of Kennedy's 13th birthday, to learn it would be a snow day. This meant both she and I would be staying home together today. I think it is a well-deserved snow day and only fitting, given the events of her original 'birth' day.

We were living outside the city at the time. It was just a few short weeks after the horrific ice storm that crippled much of Ontario, Quebec and the Atlantic provinces. It was said that the ice storm of 1998 directly affected more people than any other previous weather event in Canadian history. Like that ice storm, another January event that year would forever profoundly change the course of our lives.

Knowing how quickly I had delivered my second child, about 50 minutes start to finish, and knowing we lived about 45 minutes from the hospital, we had planned to come up to the city to stay with family within a week of my due date. Kennedy, however, had other plans. She decided to make her entrance thirteen days early.

Graeme and I had settled in to bed on the night of the 26th, where we both slept soundly until about 4:15 am, when I awoke with the need to use the washroom. Seeing the tell-tale pink when I was finished, I summoned Graeme with a, "You need to get up NOW!". We prepared ourselves quickly and made our way, excitedly and apprehensively, to the car.

Believe it or not, we both remained as calm as the still dark night we were driving through on the way up to the city, despite the dwindling number of minutes between my contractions. We made it to the hospital and my doctor was called. We were pleased he was able to make it before she was born. I believe his familiar and calm presence was one of the things that helped us through what was to come.

The labour was going very well. I was very calm, concentrating on my breathing with a musical, back and forth, conductor-like hand movement indicating the commencement and conclusion of each contraction. Graeme held my hand and comforted me throughout each one. The nurse remarked that he and I should give lessons. We all had a laugh about that. One oddity occurred that neither Graeme nor I thought much of but, I am sure, alerted knowing hospital staff that something may be not quite right. When my water broke, it kept coming. And coming. And coming. It was like Niagara Falls compared to what I'd experienced with my other two babies. It turns out this condition, polyhydramnios, aka "lots 'o' water," can be an indication that there are swallowing issues that have been present in utero.

**6:59 am.** The moment arrived. That moment that is simultaneously terrifying and exciting. It's like the feeling you get as you inch up to the top of that first incline on a rollercoaster and prepare yourself for the crazy ride that will ensue once you've reached that apex. There's no turning back now. And so, with very few pushes, our beautiful baby girl made her grand entrance into the arms of my family doctor. We held our breath and waited. Nothing. The only sounds in that small contrived-to-look-homey room were the scrambling noises of the nurse and my doctor as they whisked her over to a little table and began to work on her. To their credit, they were incredibly calm and reassuring, given the fact that the baby in front of them was cyanotic, her skin getting darker and darker before their eyes. "We just need to clear her nose out." Then, "We're going to take her down the hall to get her nose cleaned out." That was the last thing I remembered hearing.

A half hour or so later, Graeme helped me up off the bed so I could get cleaned up. I was still lying there in the same position I'd been in since she was born. I was dressed and sitting up on the bed. We called a few people to give them the wonderful news, "It's a girl!" "Yes, everything is fine, they are just cleaning her nose out. There was a lot of *vernix*. Yeah, you know, that white, thick gunk that's all over babies when they're born." As the minutes passed, though, we looked at each other, still cautiously optimistic, still blissfully ignorant, but wondering what was really going on and when they'd bring her back to us.

We eventually were taken down to a room on the floor, out of the Labor and Delivery Unit. The minutes seemed to take forever as we waited for word, all the while listening to the ebb and flow of the busy floor: nurses bringing babies from the nursery to parents, babies crying, and parents caring for them.

We finally received news sometime around noon, or maybe it was a bit after that. The day from that point on becomes a little blurry for me. It seems their attempts to pass a catheter up her nostrils to clean out her nose were futile. She had a condition called 'bilateral choanal atresia' (which was the first indication of what we would learn was CHARGE syndrome in the coming days). I remember spending much of the afternoon worrying and crying. Crying because I didn't know what the future would hold for my beautiful daughter, crying because there was something wrong that I didn't quite understand, crying because I'd given birth 6 or 7 hours ago and still hadn't seen her and crying because I couldn't even pronounce the name of the darn condition the doctor had told us.

Late in the afternoon, we finally got to go down to the neonatal intensive care unit to see her for the first time. There she was. A big, 7 pound 14 ounce baby, looking so huge in the little incubator next to all the preemies around her for whom the incubators were designed. She had a large black contraption in her mouth to keep it open so she could breathe from her mouth and was hooked up to what seemed like a zillion tubes and electrodes. Yet, despite all the hospital paraphernalia, she was our beautiful baby girl. I knew, in that moment when we first laid eyes on her, that our lives would be forever changed but I also knew that we would love her more than any words could ever express. We would go to the ends of Earth and back for her. No matter what was to come, we would get through it. And we did.

Fast forward to today: it is another January 27th and we have reached yet another amazing milestone. Kennedy is a **teenager**. She has survived 20 surgeries, hundreds of appointments and tests, hours and hours and hours of therapy and remains one of the happiest people I have met in my entire life. She is a cheerleader. She is an actor. She sings and dances. She gives speeches. She runs and jumps. She loves movies. She loves games. She loves the Wii. She loves Facebook and chatting with friends on the computer. She loves Justin Bieber and Glee. She loves her cell phone and her iPod. She has done so much more than we could have ever possibly dreamed on that very first day. She has an exuberance for life and energy that would lift the spirits of even the most cantankerous curmudgeon.

And so here we are. It is a snow day. I am a teacher and Kennedy is a student. We will be **together** all day today, which I feel is most deserving, considering the amount of time we spent **apart** on her very first day.

It is truly a *gift* to be her parents. She makes us happy every day.