

CHARGE Syndrome Foundation, Inc.

573-499-4694 (voice/fax)
1-800-442-7604 (families)
www.chargesyndrome.org



Dear Parents,

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

This letter comes to you from the CHARGE Syndrome Foundation. We are an organization of parents and professionals who want to make dealing with CHARGE syndrome a little easier by offering support, information and other services. We are here to support you as you enter into this new and challenging phase of life involving parenting or caring for a child with CHARGE syndrome.

This is a time to celebrate the new addition to your family! This new life will bring joy to your family that you didn't know was possible. Your baby is a baby first with his/her own unique personality and talents. Your baby will reach each new milestone when he/she is ready. With these kids, the hurdles may take much longer to overcome but they will work harder than most and bring such excitement as you watch them succeed. Never underestimate their abilities.

We would like to provide you with information and support to start you on your new and, at times, challenging journey. Please contact us at (800)442-7604. You can leave a message anytime. We will be happy to send you more information, invite you to join the Foundation and receive our mailings, and/or put you in touch with other parents who have been through what you are going through now.

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 for just being him/herself.

We look forward to hearing from you soon!!

The CHARGE Syndrome Foundation

February, 2005

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Who Are We?

We, the CHARGE Syndrome Foundation, Inc., are an organization of parents and professionals which grew out of a desire to help make life a little easier for families dealing with CHARGE syndrome. The Foundation is a non-profit organization providing information to families and professionals involved with individuals with CHARGE. To offset newsletter and other expenses, we have annual membership fees as follows:

Family: \$15, Professional: \$20, Organizations, Institutions and Libraries: \$30. Included with the Foundation membership is our quarterly newsletter, *CHARGE Accounts*. You can see information about the Foundation on our website www.chargesyndrome.org. You can contact the CHARGE Syndrome Foundation by calling (800)442-7604 or by email marion@chargesyndrome.org. You can leave a message anytime.

The services provided by the Foundation include:

Public Awareness/Materials:

- **The CHARGE Syndrome Foundation Website**, at 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 parent's page, deafblind resources, a listserv for families and professionals to share advice or opinions, and other resources. You can also see old issues of the newsletter online.
- **CHARGE Syndrome: Management Manual for Parents** (version 2.1, 2002, 275 pages, \$25). This is an "interactive" publication and a must for every family (and useful to professionals as well). There are nearly 150 pages of medical information for both families and professionals, more than 100 pages of developmental and educational information, a 15-page glossary, and a listing of resources. The three ring format allows for updating and personalizing. The manual is a work in progress and will be updated as the need arises. The Manual is also available for download at the website. A Spanish version of the manual (print only) is available
- **CHARGE Accounts** is a quarterly newsletter sent to all members of the Foundation. It is primarily for parents of individuals with CHARGE syndrome. The newsletter includes articles related to CHARGE, letters from families, photographs of children, resources, and calendars of special events, and Foundation news.

- The CHARGE Syndrome Medical Bibliography is a listing of medical articles on CHARGE. The listing consists of references only, not the actual articles. (cost: \$4.00)

Parent Support:

- The information line, (800)442-7604, is available to parents for information, support and/or finding other families in your area. A parent-to-parent contact list is available on request.

Biennial Conference:

- The International CHARGE Syndrome Conference is held every two years. The purpose of the conference is to bring together a large number of families and professionals who have a common interest in CHARGE syndrome in order to gain the latest medical, educational, and advocacy information available on CHARGE syndrome and to share experiences for mutual support. There are scholarships available to assist those families attending their first conference. Please contact the Foundation office for an application or more information.

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What Are The Facts About CHARGE Syndrome??

- ▶ CHARGE syndrome refers to individuals with a specific set of birth defects and medical problems.
- ▶ CHARGE syndrome is a genetic condition. The first gene for CHARGE (CHD7) was discovered in 2004. It is usually sporadic with no other affected individuals in the family. There are rare families with multiple affected individuals. CHARGE syndrome is not caused by any known exposures during pregnancy. It is not related to sex, race, nationality, religion or socio-economic status.
- ▶ There is a DNA test (CHD7) that can confirm the diagnosis of CHARGE syndrome in many cases. Because not all people with CHARGE have a detectable DNA change, CHARGE is still primarily a clinical diagnosis based on physical features. The diagnosis should be made by a medical geneticist who is familiar with CHARGE syndrome.
- ▶ The incidence of CHARGE is about one in every 8-10,000 births. The frequency is the same in males and females.
- ▶ There is a wide variation in the physical and mental abilities among individuals with CHARGE.
- ▶ Although there are many problems, children with CHARGE can survive and become healthy, happy citizens.
- ▶ Appropriate therapies and educational intervention must take into account any hearing and vision loss which is present. The intelligence of children with CHARGE is often underestimated due to the combined hearing and vision issues.
- ▶ Individuals with CHARGE need supportive, loving homes, early intervention, appropriate and challenging educational and vocational programs, and preventative medical care.

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Who? What? Where? When? Why? Frequently Asked Questions

You have been told that your baby has or may have a condition called *CHARGE* syndrome. Your baby probably has several medical complications 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 us for advice and support. Here are some questions and responses from other parents dealing with *CHARGE*. Keep in mind these are responses of PARENTS, not doctors. They are not a replacement for your doctor's advice.

1) Why do they think my child has *CHARGE*?

Your child probably has several features that are unusual in other conditions but common in children with *CHARGE*. These may include coloboma, choanal atresia, unusual ears, or other problems. Your child has probably had several tests, such as a chromosome test, to rule out other conditions. A DNA test may confirm the diagnosis of *CHARGE* in some cases, but *CHARGE* is still diagnosed primarily based on clinical features. If your child has been examined by a Medical Geneticist, he or she should be able to tell you why the diagnosis of *CHARGE* is being considered.

2) How does *CHARGE* Syndrome happen? Will it happen again?

A "syndrome" is a recognizable pattern of birth defects or other features, often with a recognized cause (single gene or chromosome abnormality, for example). Recent research has shown that many cases of *CHARGE* are caused by a new 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. For parents with one child with *CHARGE*, the recurrence risk is low but it is not zero. It is probably around 1-2%. There are only a handful of documented examples of more than one child with *CHARGE* in a family.

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

Most children with *CHARGE* have problems in many organ systems (eyes, heart, kidney, etc.) Therefore many different specialists are involved. Unfortunately, the communication between all these specialists and with the parents is often less than

ideal. It is very helpful if you can identify one specialist who is in charge of coordinating your child's overall care. It may be a cardiologist, a geneticist, or someone else. Ask if you can have a "team meeting" to bring together all the specialists to explain to you and to each other the plan for care of your child.

4) What specialists should we see?

This depends on the medical involvement of your child. All children in whom CHARGE is suspected should have examinations by cardiology (heart), ophthalmology (eye), audiology (hearing), and urology (kidney ultrasound at least) to determine which problems your child has and does not have. Depending on what is found, many will also be involved with ENT (ear, nose and throat), cleft palate team, feeding team and /or other specialists. If a geneticist is involved, he or she may be helpful in coordinating the various team members and helping you understand the roles of the different "-ologists". 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. They need to be seen by a pediatrician for all of the regular things like immunizations, weight checks, and ear infections. Your pediatrician can be an advocate for you and your child in the complex medical systems.

5) Will my child see or hear?

In the early stages of a newborn's life, it is difficult to have accurate testing or be able to predict eventual vision and hearing abilities. Many doctors will give parents a worst case scenario. Early diagnosis/prognosis could be inaccurate. Routine visits to a pediatric ophthalmologist, ENT(ear, nose and throat doctor) as well as an audiologist will help uncover your child's abilities. But you as parents or caregivers know your children best. Keep the doctors informed of the progress you see at home. Remember doctors see your children for short periods of time outside of the home setting. You live with them. Most of our children do have limited vision and/or hearing - the key word being limited. Take comfort in knowing that these kids learn naturally to compensate by using whatever vision or hearing they have, because it is all they know.

6) How does CHARGE affect my child's cognitive abilities?

The sensory losses and time lost to surgeries and frequent illnesses have a huge effect on the child's exposure to the stimulation that will shape the child's cognitive ability as well as other skills. We expect children with complex medical issues to be delayed, but that doesn't mean catch-up won't happen. Because of the sensory deficits, especially vision loss and hearing loss, communication is a big concern. It is not possible to determine cognitive ability before a communication system has been established. Often our children's intelligence is underestimated due to vision, hearing, learning, motor, and/or speech disabilities. Take advantage of all the

services available and help your child reach his/her full potential regardless of what that may be.

7) What additional services do you need?

Most states have Early Intervention programs available for children from birth to age three and school-based programs from 3 to 5. Most Early Intervention 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.

Many of the programs are paid for by the state and offer services at no charge for children who qualify. In some states, there are costs or fees. If you have not been put in touch with your local program, contact your local school district or mental health/mental retardation programs to find out what is available in your area.

In the US, every state has a DeafBlind coordinator. Your child does not have to be deaf or blind to qualify. If he or she has vision loss and hearing loss, he or she may qualify for services through the Deafblind program.

If you have not heard of these programs yet, ask to speak with a hospital social worker and ask him/her to help you find out more about what is available in your area. You may want to look into programs while your child is still in the hospital to get services started as soon as possible.

8) What does my child's future look like?

Although these children have many problems, they can survive and become healthy, happy citizens. Doctors' visits and medical problems taper off and/or change as your child grows. Keep in mind, every child is different. Accept not knowing and enjoy today while doing what is possible to prepare for the future. NEVER underestimate your child's abilities. Always put the disability second. Be involved, interact and enjoy because, as hard as it may be at times, they grow up fast, overcome many obstacles, and will make you proud.

9) How can I find out more about CHARGE?

- Join the CHARGE Syndrome Foundation. The cost is \$15 per year. If you cannot afford it right now, the fee will be waived, just let us know.
- If you have Internet access, check out the Website www.chargesyndrome.org.
- If you have email, consider joining the listserv (see link at the Website).
- Order the CHARGE Syndrome: Management Manual for Parents. It's only \$25 and it has many pages of useful information, medical, developmental, and educational. It will also help you organize your child's medical records.

Any other questions?? Please contact us at the Foundation and we'll do our best to help.

One Family's Story

by Michelle Westmaas

I recall our devastation and bewilderment at Aubrie's birth. Her medical needs seemed huge. She had heart surgery at 8 days old and remained hospitalized for her first few weeks of life. She could not eat orally. We were told that she may be deaf-blind and mentally retarded. When she was first released from the hospital, we were on a 2-hour, round-the-clock feeding schedule. I was pumping breast milk, and feeding it to her slowly via a nasogastric tube, then watching her carefully to assist when she vomited. Almost immediately, it was time to start the cycle again. We were exhausted and overwhelmed. Soon, she was back in the hospital with failure to thrive and we were at the end of our physical, mental, and emotional resources.

I can't remember the time frame of those next few months. Somewhere in there, we fought our insurance company to get overnight home nursing care. They gave us just a few nights a week of relief. We had health department nurses checking her weight and basic health status regularly. We traveled two hours each way to the Children's Hospital for therapy and doctor appointments at least 2-3 times each week. It was crazy. My older son was in kindergarten at the time (6 years old) so my husband tried to keep his life as regular as possible while I ran myself ragged keeping up with Aubrie's needs.

It wasn't long before we had therapists from all areas (hearing, vision, deaf blind, etc) coming to the house several days a week. We continued to make the trip to the Children's Hospital for speech and feeding therapy. We were making that trip once or twice a week for over a year. That's the bleak picture of Aubrie's infancy. I remember my friends coming around trying to show support. They'd say, "I just don't know what to say." My reply was that I didn't know what they could say either. It was helpful to me that they obviously cared. They would share my tears and concerns. They helped bear the load by caring for my son when I had to be away, providing meals when I didn't have the energy to worry about feeding my family or myself, accompanying me on trips to the hospital so I wouldn't always have to be alone. We live in a small rural community. The outpouring of support and empathy was incredibly helpful to us.

Some things that stand out in my mind -- At one point, the local pastor came for a visit. I am not a church-goer, but many of my friends are in his congregation and had expressed their concern for our family. One thing he told me that I'll always remember -- "Allow your friends to help you. Give them the gift of being able to help and do something for you." I was trying to do it all because I didn't want to impose on my friends. But he helped me see that there was a large group of people who cared so much and yet didn't know how to help. It was a comfort to them when I would let them know what they could do to help me.

One day I was just so tired I couldn't go on. I knew I had to sleep. But Aubrie needed constant supervision. I called friends and all were busy. They called upon people they knew, and before long, a kind woman from the community was at my house sitting with my very ill baby while I slept upstairs. I could not have gone on without that rest. What an angel she was!

The therapists that came to attend to Aubrie's hearing, vision, and medical needs were just as critically therapeutic to me. They would listen to my repeated questions and concerns. They would allow my tears and be with me. They would give me the tools I needed to care for my child. It was frustrating to have a baby that I didn't know how to care for -- how could I not know how to feed my baby? I had to learn how to manage the tube feedings. And why would my baby not eat?? What if she couldn't hear or see me?

I remember clearly the first time she responded in joy when I entered a room. She was about 4 months old, I think. That's a long time to wait for your baby to recognize you and respond. The baby's response to Mom -- that "mother-child dance" -- is a huge part of infancy for both the child's development and for the mother. It's what rewards the mom for all of her dedication. It's difficult to talk and coo at a baby that doesn't seem to hear or see you. You feel sort of foolish when you are the only one engaging in the "dance". However it is critical to continue. I recall singing lullabies to Aubrie with her held tight up by my cheek. I wanted her to feel the rhythm of my voice and my breath on her cheek in case she couldn't hear me. She didn't even like to be held at first. That was heartbreaking. We purposely held and cuddled her as much as possible even if she didn't seem to like or need it. However, when she was distressed and needed to comfort herself, I always allowed her that. And I even had to tell Grandma sometimes to leave her alone to cry and comfort herself. It goes against your instincts to not pick up a crying distressed infant.

I felt so different suddenly from my friends. I felt somehow inferior too. I needed my friends to continue to value me as the person I was before I became "the mother of a child with special needs". The CHARGE listserv was a lifeline for me (ed. Note: see information sheet or link at www.chargesyndrome.org). It was the only way that I could feel a part of a community and a family. Without that, I would have felt so isolated. The families on the list were there for me when I needed emotional support.

I also learned about what I might expect for Aubrie's future. No one can predict the future of our kids because the syndrome affects each differently. However, at least I felt like I had a small glimpse into the future. It seems that one of the painful things for new parents is not knowing what the future will be like for their child. When you have a baby, you have dreams and visions for that child. You can imagine them someday riding a bicycle, having a birthday party, having their best friends over to spend the night, going on dates, driving a car... With a baby with CHARGE, suddenly all of those dreams and visions go up in smoke. Will this child ever ride a bike? Now you are wondering about things you always took for granted -- will this child eat? Will this child see? Will this child speak?

There is no help for that dilemma. It takes time for new parents to come to terms with the uncertainty, to gain new knowledge that allows them to build new hopes and dreams, and to learn to love their child absolutely unconditionally. Again -- just being supportive, listening to and understanding their concerns -- that's all friends and family can do.

Now -- let me tell you about our life today. Aubrie at age 5 is eating, walking, running, talking, singing, beginning to read!! At age 2 or 3, my greatest hope was that she would be mobile somehow by kindergarten -- I would have been happy if she could crawl into the classroom. I hoped she'd be able to eat or at least

appear to eat normally with her peers at snack time even if she had to come home for tube feedings at night. I never would have allowed myself to imagine what she has really become.

She will go to kindergarten next year. She is in her 3rd year of preschool. She can maneuver herself on the playground independently. She recites her nursery rhymes, sings favorite songs, argues with her brother, and plays cooperatively with her friends... She can eat anything -- chicken legs, pizza, and licorice. She knows all the letters of the alphabet and their sounds and is beginning to recognize words by sight. She can write her name and tries to write other words with invented spellings. She is invited to friends' birthday parties. She has friends over to play and is invited to play at other kids' houses. She takes dance class with her peers with no special assistance. She uses the toilet independently, gets herself dressed and undressed, puts on her own coat and shoes. She makes microwave popcorn all by herself. Her speech is unintelligible to people who don't know her -- and sometimes to us at home too. But it's coming along. Her writing skills are weak -- but she's learning and she's doing it! Her balance is a bit off -- but she manages. I put on her glasses and hearing aids, but she insists on turning them on herself. She's an amazing little girl! She's truly our princess.

Aubrie has very, very limited vision in one eye, but near normal in the other. She has mild-moderate hearing loss in one ear, and moderate-severe in the other. With her glasses and hearing aids, she sees and hears in the normal range. She has issues in all areas of development, but all of them are relatively minor.

So... although it is incredibly difficult in infancy, things do get better. Not all of the children are doing as well as Aubrie by age 5. Each child is different. But I would venture to say that most families are doing well by age 5 or 6. Regardless of the severity of the child's problems, the family will adjust to the new "normal" of their lives. I never thought I'd have a life or an identity of my own again -- but I have my own business now! There's another mom from the list who was equally as overwhelmed as I was in the beginning. Her child is now 6 or 7, more involved than Aubrie, but doing well -- and Mom is working again, taking care of herself physically by exercising and running in 5K races. Both of us used to wonder if there was ever a light at the end of the tunnel. We certainly could see no evidence of it. But it came! Now I think of it as a very winding tunnel. The light is at the end, but you have to get around many, many bends before you can see it.

New parents need to know that these difficult times will improve. They may not be able to see how or have any reason to believe that it will. They may not be able to imagine how this will ever get better or be ok, but they need to just have faith and know in their hearts that it will be ok and they will get through this. If they have computer access, they can join the listserv. Having a connection to the greater CHARGE family can be very comforting and helpful.

A book that I found therapeutic is "Changed by a Child" (Doubleday, 1997). I would read it each night and cry, cry, cry. But it helped me to understand what this world was that I was entering. I was a person that craved information. There is a poem called, "Welcome to Holland". It describes the feeling of suddenly being in an unexpected place. I remember the other parents of children with special needs saying that their special child was a blessing and blah, blah, blah. I thought they were so full of it! How could they really

think that all the problems their children had could be a blessing!!! Then one day, I realized that I, too, could see how CHARGE has positively affected my life and that of my family and friends. Of course, if I could wave a magic wand and make CHARGE go away, I certainly would. But Aubrie has brought us a wealth of blessings that she could not have brought without CHARGE.

Another thought that helped me in the very beginning -- We knew that Aubrie was going to be a girl and we named her in utero. I even sent an email announcing the expected arrival of Aubrie Michele Westmaas! She had a definite place and identity in our family long before she was born. Then she came and she was not as expected. But I already loved her - just as I loved my son. If he had a sudden accident and became disabled, I wouldn't love him any less. The essence of who he is wouldn't change. So I looked at Aubrie in the same way. Unfortunately, I didn't get to know her before CHARGE happened. But I did love her -- the essence of her -- the whole her -- regardless of the package she came in. One person on the list had a comeback for people who stared at her child -- "You are one car accident away from my life". It is true that the "disability club" is one that any of us can become a member of at any moment without our consent. It's the only minority group I can think of that you do not necessarily join at birth. Any one of us can become disabled at any moment.

I found it very disturbing and frustrating that my own birth child would be part of a minority to group to which neither my husband nor I belong. And I had to learn a new language (sign) to communicate with her. What was that all about? How could that be right or fair? Then I found a way to reconcile those angry feelings for myself. I began to think of Aubrie as having 3 parents. She has genetic material from me, she has genetic material from my husband, and she has genetic material from CHARGE. She looks a bit like a member of our family, but she also greatly resembles her CHARGE peers (who genetically are somewhat like siblings to her!).

Since I have come to know and cherish our CHARGE family, I am comfortable sharing her with them. We even had the good fortune to meet a CHARGE "sibling" who is very much like Aubrie. I call them "twins 13 years removed". I was able to sit with this young woman at the last CHARGE conference. She is a fabulous girl. Sitting there, I felt like I finally did have a glimpse into Aubrie's future - and if this was it, if this was what it would be like to sit beside my adult daughter someday, then I was happy to embrace it.