**CHARGE Syndrome Foundation, Inc.**

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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 $25 per year for families/individuals or $40 for professionals. 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).

-Download the CHARGE Syndrome: Management Manual for Parents. It is on our website for free download at http://www.chargesyndrome.org/resources.asp. If you would like a hard (paper) copy, 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.