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, choanalatresia, 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, choanalatresia, 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 is 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.

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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 gonadalmosaicism, 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 is it 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 medial 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.