

Disability Awareness Begins With You



CHARGE Syndrome

Q: What is CHARGE Syndrome?

A: CHARGE Syndrome refers to children with a specific set of birth defects. 'CHARGE' originally came from the first letter of some of the most common features seen in children with this syndrome.

C = Coloboma and Cranial nerves

A coloboma is a cleft or failure to close of the eyeball. This can result in a keyhole shaped pupil and/or abnormalities in the retina or optic nerve. Colobomas of the retina or optic nerve may result in significant vision loss, especially visual field defects in the upper half of the visual field. Visual acuity may also be affected, resulting in nearsightedness or farsightedness. Surgery cannot correct ocular colobomas, but glasses often help with visual acuity. Children with CHARGE Syndrome are often very sensitive to light. Many are more comfortable with sunglasses, even indoors.

Due to cranial nerve problems about 40% of children with CHARGE Syndrome have facial palsy, and at least 30% have swallowing problems. Children with facial palsy may be more likely to have sensorineural hearing loss. Swallowing problems often last for years, but typically resolve by age 7 or 8 years.

H = Heart

About 80% of children with CHARGE Syndrome are born with a heart defect. Many are minor defects, but many require treatment or surgery.

A = Atresia of the choanae

The choanae are the passages from the back of the nose to the throat which make it possible to breathe through the nose. In children with CHARGE Syndrome, these passages may be blocked (atresia) or narrowed. Surgery can often correct these defects.

R = Retardation of growth and development

Although most children with CHARGE Syndrome are average size at birth, many will become small due to nutrition problems, heart problems, or growth hormone deficiency. Some catch up after the severe medical problems and feeding problems have resolved. Most children with CHARGE Syndrome will be developmentally delayed. Often, this is primarily due to sensory deficits (vision loss and/or hearing loss) and frequent hospitalizations as infants. Some children with CHARGE Syndrome will be mentally retarded, with or without brain abnormalities.

G = Genital and urinary abnormalities

Many boys with CHARGE Syndrome have a small

penis and/or undescended testes. Girls may have small labia. They may require hormone therapy to achieve puberty for either sex. They may also have kidney or urinary tract abnormalities, especially reflux.

E = Ear abnormalities and hearing loss

Most children with CHARGE Syndrome have unusual external ears, including short, wide ears with little or no earlobe, often with a "snipped off" appearance to the outer fold of the ear. The ears may be soft due to floppy cartilage. Keeping a hearing aid in place is often difficult. Hearing loss (conductive and/or nerve) is present in 80 - 85% of children with CHARGE Syndrome, ranging from mild hearing loss to profound deafness. Many children have additional conductive losses due to frequent ear infections and also have difficulty with balance. Children with CHARGE Syndrome may have other birth defects, including cleft lip and palate and have weak upper body strength.

How often does it occur?

The incidence of CHARGE Syndrome is about 1/10,000 births. It is probably very underdiagnosed. The frequency is the same in males and females. CHARGE Syndrome has been seen in all races. CHARGE Syndrome is usually sporadic with no other affected individuals in the family. There are rare reports of multiple affected individuals in a family. Recurrence risk is low, probably 1-2%. The risk of an affected individual having an affected child may be much higher.

What therapies/helps exist?

Although these children have many problems, they can survive and become healthy, happy citizens. In any child in whom CHARGE Syndrome is suspected, complete cardiac, eye, and hearing evaluations should be performed, as well as abdominal ultrasound, and chromosome evaluation. The intelligence of children with CHARGE Syndrome is often underestimated due to the combined hearing and vision problems. Management should be by a multidisciplinary team and coordinated by a single person, if possible. Many of the effects of CHARGE can be corrected with surgery. Major problems of CHARGE such as atresia of the choanae and heart problems often can be operated on for some correction. Steroids can be administered to regulate/correct problems with reproductive organs. Also other abnormalities often associated with CHARGE Syndrome such as cleft lip and/or palate, difficulty swallowing, and seizures can be controlled with

surgery and/or medication. Patients can also use hearing aids and glasses to aid with vision or hearing loss.

Children may be physically very frail due to the effects of CHARGE Syndrome on the body, interfering with the cognitive development of the child. Children may be in and out of hospitals constantly for the first few years of life. Often children may go undiagnosed due to lack of knowledge about the disease. To be diagnosed with CHARGE Syndrome the child must display at least four of the six major characteristics associated with CHARGE Syndrome.

Teachers and team members must be aware that it may be appropriate for the child to wear sunglasses during times when there is bright light. As a result of the coloboma the eyes may be highly sensitive to light. Motor coordination may be affected as a result of poor vision because the child is not able to see what other things are going on around him/her. Teachers should be sensitive to presenting information from different angles that the child may see more. The child may have a hearing loss that is sensorineural, conductive, or mixed with varying degrees of loss. The child with CHARGE may be "hampered" because of the inability to communicate because the hearing loss has caused a language delay. The sooner that all "deficits" are detected the earlier the team can begin to address the situation and teach the child.

Other Insights:

In an educational setting all involved must be aware of the special needs a child with CHARGE may have. Because CHARGE can affect the eyes, ears, and brain it is most important that all members of the educational team take each of these factors into account. Teachers also must be aware of how to teach parents how to work with their children. Parents are often in shock when their child is diagnosed with one debilitating problem. With several areas of disability families can be much more traumatized. It is imperative that teachers remain sensitive to parents and families.

For more information:

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