

What is CHARGE Association?

CHARGE Association (or Charge Syndrome) refers to a specific set of birth defects. The word “CHARGE” comes from the first letter of some of the most common features seen in children with these problems. Distinguishing between CHARGE Association and other conditions should be done by an experienced medical geneticist.

Characteristics

C

Coloboma/Cranial Nerve Abnormalities

A coloboma is a cleft in the eye, retina or optic nerve. This can result in a keyhole-shaped pupil and/or abnormalities in the formation of the retina or optic nerve. Colobomas of the iris may limit the child's *ability to adjust to bright light*. Colobomas of the retina or optic nerve may result in significant vision loss, including *blind spots in the visual field*. Colobomas cannot be corrected with surgery. Eyeglasses may help correct visual acuity (near or far sightedness), but cannot help with visual field defects (blind spots). Many children with CHARGE have cranial nerve abnormalities involving the nerves which supply the muscles of the face. These cranial nerve problems may lead to *weakness in the facial muscles and swallowing difficulties*.

H

Heart Malformations

At least two-thirds of the children with CHARGE are born with heart defects. Some are minor defects, but many require treatment or surgery. Some of the more complex heart defects seen in CHARGE, such as tetralogy of Fallot, may be life threatening.

A

Atresia of the Choanae

The choanae are the passages from the back of the nose to the throat which makes it possible to breathe through the nose. These passages may be blocked (atresia) or narrowed (stenosis) and may require surgery. Many young children with CHARGE experience *apnea, difficulty coordinating breathing and feeding, or other breathing problems*.

R

Retardation of Growth and/or Development

Some children with CHARGE may be *small* due to nutritional problems, heart problems, or growth hormone deficiency. Many of the children will be *developmentally delayed*. Often, this is due primarily to sensory deficits (vision and/or hearing loss), especially when coupled with frequent hospitalizations as infants. Some children with CHARGE will be cognitively delayed, with or without brain abnormalities. Cognitive function appears to be on a continuum.

G

Genital and/or Urinary Abnormalities

Many boys with CHARGE have small penises and/or undescended testes. Boys or girls may require hormone therapy to achieve puberty. Children with CHARGE may also have kidney or urinary tract abnormalities, sometimes requiring surgery.

E

Ear Abnormalities/Hearing Loss

Ear anomalies can affect the external ear (which may be unusually large or small or of an unusual shape), middle ear (bone malformations or chronic infections), and/or the internal ear (especially high frequency hearing loss). The most common form of hearing loss found in children with CHARGE Association is mixed, that is a conductive loss because of middle-ear problems combined with sensori-neural loss because of problems with the cochlea. Some children with CHARGE may have central auditory processing problems (difficulties processing auditory information in the brain) which are very difficult to detect and can be superimposed over the other hearing problems.

There are no consistent chromosome abnormalities, environmental factors, or pregnancy exposures known to cause CHARGE Association. There are rare reports of multiple affected individuals in a family (siblings, or parent and child). If there is no one else in a family affected, the recurrence rate is low, probably around 1-2%. However, the risk of an affected individual having an affected child may be much higher. The incidence of CHARGE is about 1/10,000 births, but it is probably very underdiagnosed. The frequency of occurrence is the same in males and females and CHARGE has been seen in all races.

Other Features

Some children with CHARGE may have other problems such as: cleft lip and palate, tracheo-esophageal fistula or atresia, poor immune response, weak cartilage in the ears and trachea, delayed and abnormal dental development, hypoglycemia or low blood sugar, and malformed or absent semi-circular canals which may affect the sense of balance.

Treatment/Prognosis

Although there are a number of medical problems seen with CHARGE Association, most children with CHARGE survive and become healthy, happy, and productive citizens. Medical and surgical procedures that correct malformations and establish the health of the child are important in the first year or two of life, but equally important are developmental issues. A team approach utilizing physicians, educators, and therapy personnel to provide early intervention to the child and the family is critical to obtaining optimal developmental outcomes for the child. Especially if vision and hearing loss are both present, educational programming from specialists in deafblindness will also be an important and necessary program component.

Resources

CHARGE Syndrome Foundation, Inc., 2004 Parkdale Blvd., Columbia, MO 65202, 800-442-7604 or 573-499-4694, <http://www.chargesyndrome.org>.