

## What is CHARGE Syndrome?

CHARGE syndrome is a complex genetic condition seen in about 1 in 10,000 births. The name "CHARGE" is an acronym stemming from some of the features seen in many children with CHARGE: **C**oloboma, **H**ear, **A**tresia of the choanae, **R**etardation of growth and development, **G**enito-urinary, **E**ar anomalies and hearing loss.

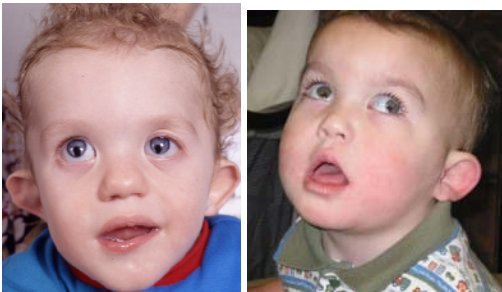


Infant with gastrostomy tube for feeding

CHARGE syndrome is no longer diagnosed using the acronym features. A diagnosis of CHARGE syndrome is made using **Major** and **Minor** clinical features or by finding a mutation in the CHD7 gene.

**Major features** of CHARGE syndrome are:

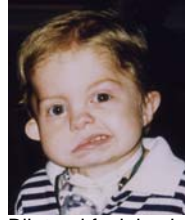
**Coloboma:** Coloboma is a cleft of the eyeball. It can result in a keyhole-shaped pupil (iris coloboma) and/or abnormalities in the retina or optic nerve. Colobomas of the retina or optic nerve can cause vision loss or blindness.



Iris coloboma (also note typical ears and facial palsy)

**Choanal atresia or stenosis:** a blockage or narrowing of the airway from the back of the nose to the throat. Many individuals with CHARGE also have apnea or other breathing problems.

**Cranial nerve abnormalities.** Most individuals with CHARGE syndrome have abnormalities of the cranial nerves (nerves for the head and neck), especially facial palsy (partial paralysis of the facial muscles), swallowing difficulties, and decreased or absent sense of smell. The swallowing problems can be severe and require tube feeding for many years to get adequate nutrition and avoid aspiration pneumonia.

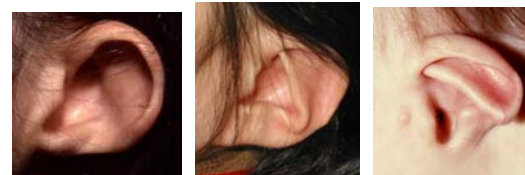
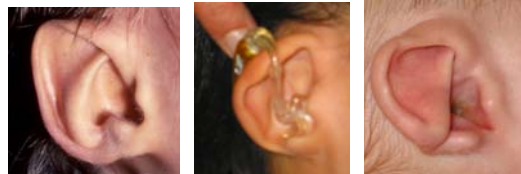


Bilateral facial palsy and tracheostomy



Gastrostomy (g-tube) feeding

**Characteristic CHARGE ears:** Most individuals with CHARGE syndrome have unusually formed ears. They are often short, wide (cupped) ears with little or no earlobe. Sometimes the outer fold is missing. They are often very floppy.



Typical CHARGE ears

Most people with CHARGE syndrome have **hearing loss**, ranging from mild to profound and inner ear problems leading to **poor balance**.

**Minor features** of CHARGE syndrome are often helpful in making a diagnosis. They include heart

defects (often complex and difficult to correct), cleft lip or palate, Tracheo-esophageal (TE) fistula or esophageal atresia, genital abnormalities, kidney problems, characteristic face, and hockey-stick hand crease.



Hockey stick crease



Typical CHARGE face



Teen face



Young adult face

Other features of CHARGE syndrome may not be as helpful in making a diagnosis, but they can be very important for management. These include growth deficiency, poor upper body strength, sleep difficulties, and typical behaviors (perseverative behavior, obsessive-compulsive disorder).

## What causes CHARGE syndrome?

CHARGE syndrome is a genetic condition most often caused by a new change (mutation) in the CHD7 gene on chromosome #8. It is usually sporadic, with no other affected family members. It is found in every ethnic group all over the world.

## What else can look like CHARGE syndrome?

Other conditions with features similar to CHARGE syndrome include velocardiofacial (VCF) syndrome (also called 22q deletion syndrome), VATER/VACTERAL association, and prenatal retinoic acid (Accutane) exposure. Evaluation by a clinical geneticist and lab tests can help determine which condition is present.

## Development in CHARGE syndrome

Children with CHARGE syndrome have developmental delays. Some will have mental retardation, but many have normal intelligence. Because of all of the medical issues, many children spend months in the hospital and have multiple surgeries, especially in the first few years of life. Most are followed by six or more medical specialists. Even so, referrals to educational resources as early as possible are very important. Children with both hearing loss and vision loss are classified as "deafblind." These children need to be followed by specialists in deafblind. **Early diagnosis and intervention for hearing loss and vision loss are critical to development of communication.**



Kids enjoying sun and pool at Miami conference, 2005

Children with vision and/or hearing problems experience the world differently. The intelligence of children with CHARGE is often underestimated due to the combined vision and hearing loss and the multiple medical issues. Each child's specific hearing and vision status must be taken into consideration when designing infant stimulation, physical therapy, and speech and language programs. A comprehensive educational program should include information on balance, muscle tone, breathing, feeding, and other specific needs, including vision, hearing and communication.

CHARGE is medically and developmentally one of the most complex conditions known. Children with CHARGE syndrome are also likely to be amongst

the most truly 'multi sensory impaired' people you will ever meet, having difficulties not just with vision and hearing but also with the senses that perceive *balance, touch, temperature, pain, pressure, and smell.*



Two children with CHARGE at Japanese conference, 2003

There is no other identified sub-group within the population of people with multi-sensory impairment who have so many medical problems, of such complexity and severity, and with so many hidden or delayed difficulties, and yet no sub-group has shown such a consistent ability to rise triumphantly above these problems.



Five young adults with CHARGE Syndrome in Miami, 2005

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The CHARGE Syndrome Foundation, Inc. is a non-profit 501(c)(3) charitable organization

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# CHARGE Syndrome



We provide information on CHARGE syndrome to:

- Parents of children with CHARGE,
- Individuals with CHARGE, and
- Professionals who treat individuals with CHARGE

### Resources include:

- Quarterly Newsletter
- New Parent Packet (free)
- CHARGE Syndrome: a Management Manual for Parents
- Bibliography
- Parent to parent information
- Biennial conferences for families and professionals