

# CHARGE Syndrome



## **What is CHARGE Syndrome?**

CHARGE Syndrome is a recognizable (genetic) pattern of birth defects which occurs in about one in every 9-10,000 births worldwide. It is a completely complex syndrome, involving extensive medical and physical difficulties that differ from child to child. Usually there is no history of CHARGE syndrome in the family.

## **What are the symptoms of CHARGE Syndrome?**

Babies born with CHARGE syndrome are often born with life-threatening birth defects, including complex heart defects and breathing problems. Swallowing and breathing problems make life difficult even when they come home from the hospital. Most children have hearing loss, vision loss, and balance problems which delay their development and communication. All individuals will require medical and educational intervention for many years. Despite these obstacles, children with CHARGE Syndrome often far surpass their medical, physical, educational and social expectations.

## **How is CHARGE Syndrome diagnosed?**

While a CHARGE Syndrome gene has been discovered, the gene test is very expensive and isn't perfect- only 2/3 of people with CHARGE Syndrome have a positive gene test. Therefore, the diagnosis of CHARGE Syndrome is still clinical- based on the medical features seen in the child.

*Some major features of CHARGE Syndrome:*

- Coloboma of the eye- sort of like a cleft
- Choanal atresia or stenosis- narrow of the passages that go from the back of the nose to the throat
- Cranial nerve abnormality- missing or decreased sense of smell, swallowing difficulties
- CHARGE outer ear-short, wide ear with little or no lobe
- CHARGE inner ear-Malformed cochlea small or absent semicircular canals

*Some minor features of CHARGE Syndrome:*

- Heart defects

- Cleft lip/Cleft palate
- Kidney abnormalities
- Growth deficiency
- Palm crease

### **How is CHARGE Syndrome treated?**

Although children with CHARGE have many problems, they can survive and become happy healthy citizens. Many of the structural abnormalities can be surgically corrected. Others such as feeding problems and speech and language deficits may require years of therapy and interventions. Infants diagnosed with CHARGE Syndrome will need to be followed by a number of medical and developmental specialists, depending on their individual needs. Some of the medical specialists who often help children with CHARGE are:

- Genetics
- Cardiologist
- Audiologist
- ENT
- Ophthalmologist
- Urologist
- Endocrinologist
- Occupational Therapist
- Physical Therapist
- Speech Therapist

### **Resources**

- <http://rarediseases.org/>
- [www.audiology.org](http://www.audiology.org)
- [www.birthdefects.org](http://www.birthdefects.org)
- [www.chargessyndrome.org](http://www.chargessyndrome.org)
- [www.hknc.org](http://www.hknc.org)
- [www.letthemhear.org](http://www.letthemhear.org)
- <https://ghr.nlm.nih.gov/condition/charge-syndrome>
- <http://www.webmd.com/children/charge-syndrome-10769>