What else can look like CHARGE syndrome?

There are many other conditions with features that overlap with CHARGE, including 22q deletion, Kabuki and VATER. Evaluation by a clinical geneticist and lab tests can help determine which condition is present.

What happens to children with CHARGE?

CHARGE is medically and developmentally one of the most complex conditions known. There is no other identified 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.

Yet, no other group has shown such a consistent ability to rise triumphantly above their challenges than individuals with CHARGE syndrome.

Development in CHARGE syndrome

Children with CHARGE have developmental delays. These may be due to:
- Medical issues
- Sensory deficits (hearing, vision and balance disturbances)
- Brain/intellectual differences

Many have normal intelligence, but most children and adults with CHARGE will need life-long assistance due to their sensory deficits regardless of their intellectual level.

Deaf-Blindness

Most children with CHARGE are not totally deaf or totally blind. However, children with any combination of some hearing loss and some vision loss are classified as "Deaf-Blind." These children need to be evaluated by specialists in this area.

Early diagnosis and intervention for hearing loss and vision loss are critical to development of communication. Deaf-Blind resources can be found through the National Consortium on Deaf-Blindness www.nationaldb.org

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WHAT IS CHARGE SYNDROME?

CHARGE is a complex genetic syndrome that may be obvious at birth due to the combination of birth defects and medical features. It also affects the senses and behavior.

Some of the medical features
- Heart defects
- Cleft lip and/or palate
- TE Fistula or Esophageal atresia
- Choanal atresia
- Facial palsy
- Swallowing problems
- Breathing problems

Some of the sensory features
- Vision loss
- Hearing loss
- Balance problems
- Behavior issues
- Sleep disturbance

WHAT CAUSES CHARGE SYNDROME?

How often does it happen?

- CHARGE is a genetic condition, with mutations in the CHD7 gene on chromosome 8 being the major cause
- Sporadic – there is usually only one person in a family with CHARGE
- Occurs in about 1/8,000 births

How is CHARGE different from other syndromes?

Features that are common in CHARGE and rare in other conditions include:

- Coloboma of the eye
- Cranial nerve anomalies, including facial palsy, swallowing problems and lack of sense of smell
- Unusual ear shape
- Choanal atresia - blockage at the back of the nose
- Absent or abnormal semi-circular canals - balance problems
- Abnormal cochlea - a type of hearing loss

Many babies and children with CHARGE have physical features in common and sometimes look a bit like each other.