

Charge Syndrome

BIRTH DEFECT RESEARCH FOR CHILDREN



What is CHARGE Syndrome?

CHARGE is an acronym used to represent the major symptoms of this condition.



Charge Syndrome



C- coloboma (an eye defect resulting in a keyhole shaped pupil and/or abnormalities in the retina or optic nerve).
H- heart disease
A- choanal atresia (blocking or narrowing of the breathing passages in the nose)
R- retarded growth or development,
G- genital and urinary abnormalities
E- ear abnormalities and/or hearing loss.

Frequently, children with CHARGE have other birth defects, including poor immune response, cleft palate, or behavioral problems. To be classified as having CHARGE, children must have two or more of the CHARGE defects.

How many children have CHARGE?

The incidence of CHARGE is approximately 1:10,000 to 1:12,000, but studies suggest this syndrome has been under diagnosed.

How do you know if your child has CHARGE?

Approximately 80% of children with CHARGE have eye malformations, which can include a keyhole-shaped pupil, or abnormalities in the retina or optic nerve. Any one of these can result in significant vision loss, nearsightedness, farsightedness, or extreme light sensitivity. Hearing loss is also present in 80-85% of children with this syndrome. In addition, most children with CHARGE have some form of mental retardation. Also, growth retardation can be seen in a CHARGE child at approximately six months of age.

What causes CHARGE?

In most situations, an incidence of CHARGE is isolated, with the exact cause being unknown. However, some instances of autosomal dominant and/or recessive inheritance have been reported, so genetic counseling is suggested.

How can you help a child with CHARGE?

Medical Diagnosis: Every child with CHARGE should have a thorough medical assessment as soon as possible. This should include a heart, vision, and hearing check, as well as developmental and intelligence testing. Newborns are especially susceptible to respiratory failure due to blockage of the choanae.

Treatment: Visual correction or training should be done during early childhood. In addition, hearing aids and speech and/or language therapy should be instituted as soon as possible. For children with growth defects, growth hormone replacement therapy is available.

Surgery: Surgery is generally required if there is choanal atresia, and also if there are any heart defects present.

What's in the future for child with CHARGE ?

This depends on the extent of the defects and severity of retardation. The abilities of some children with CHARGE many underestimated due to the combination of hearing and vision loss. With extensive speech and language therapy, in addition to hearing and vision aids, many children with this syndrome can function well. If choanae or heart defects are surgically corrected, many CHARGE children will have a normal life expectancy.

Fact Sheet by:

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