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My46 Trait Profile

CHARGE syndrome

Other Names: Hall-Hittner syndrome

CHARGE syndrome is a genetic condition with characteristic facial features and birth defects. It is caused by mutations in the CHD7 gene which makes the chromodomain helicase DNA binding protein 7.

Characteristics of CHARGE syndrome

CHARGE syndrome is a complex genetic condition that affects approximately 1 in every 10,000 newborns. CHARGE is an acronym for some of the features commonly seen in this condition: Coloboma (i.e., hole in eye structure causing vision loss), Heart defects, choanal Atresia (i.e., blockage or narrowing of the nasal airway), growth Retardation, Ear anomalies (e.g., characteristic external, middle and inner ear abnormalities causing hearing loss and balance difficulties). Other common characteristics found in individuals with CHARGE syndrome include: cranial nerve dysfunction, decreased sense of smell, swallowing/feeding difficulties, and facial palsy. Other birth defects seen in CHARGE syndrome include cleft lip/palate, tracheoesophageal fistula (i.e., an abnormal connection between the trachea and the esophagus), kidney abnormalities, genital abnormalities, and typical behavior patterns including obsessive compulsive disorder. Some individuals with CHARGE syndrome have distinctive physical features including a square face, broad forehead, and unusually shaped ears. CHARGE syndrome is highly variable in terms of presentation and severity of symptoms. At the mild end of the spectrum, a few individuals have only hearing and balance problems.

Diagnosis/Testing

CHARGE syndrome can be diagnosed either by meeting certain clinical criteria or by genetic testing for a change or mutation in the CHD7 gene. Most individuals (65-85%) with CS have a mutation in the CHD7 gene. This gene makes the chromodomain helicase DNA binding protein 7 protein that regulates developmental processes throughout the body. When the gene has a mutation, normal development does not occur and results in the features seen in CHARGE syndrome.

Management/Surveillance

Individuals with CHARGE syndrome experience physical and developmental delays and usually require extensive medical and educational intervention. Medical intervention may include surgical correction and management of birth defects, feeding therapy, speech-language therapy, and occupational therapy. Specialized education for combined hearing and vision loss is usually required. Despite these issues, children with CHARGE syndrome often far surpass their medical and educational expectations.

Mode of inheritance

CHARGE syndrome is inherited in an autosomal dominant pattern. This means inheriting one CHD7 mutation is enough for an individual to be affected and show signs of CS. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child. Indeed, most cases of CHARGE syndrome are sporadic with no other affected individuals in the family.

Risk to family members

The risk to family members depends on whether or not the individual with CHARGE syndrome has a parent affected with CHARGE syndrome. If a parent also has CHARGE syndrome, the risk of having a child with CHARGE syndrome is 50% with each pregnancy. If a parent does not have CHARGE syndrome, the risk of future pregnancies being affected is very low.

Special considerations

None

Resources

The CHARGE Syndrome Foundation
http://chargesyndrome.org
Genetics Home Reference: CHARGE syndrome
http://ghr.nlm.nih.gov/condition/charge-syndrome

References

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