Management of CHARGE syndrome: An Issue of Great Complexity

IJSN
Volume 3, Number 2

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Abstract

CHARGE syndrome was initially defined as a non-random association of anomalies. In 1981, an expert group defined the major (the classical 4 c"s: Choanal atresia, coloboma of the eye, characteristics of ears, cranial nerve anomalies) and minor criteria CHARGE syndrome. Individual with all 4 major characteristics or three major and three minor characteristics are highly like to have CHARGE syndrome. It affects approximately 1:10,000 births world wide. Recently (2004) researchers have discovered a genetic link, specifically, a strong association between the CHARGE phenotype and a mutation of the CHD7. The official name of this gene is "chromadomain helicase DNA binding protein 7" and CHD7 is the gene's official symbol. The Children with CHARGE syndrome requires intensive medical management as well as numerous surgical interventions.[1]

Keywords: CHARGE; Choanal atresia; Coloboma.

Introduction

CHARGE syndrome is a challenging genetical disorder which affects many areas of the body. It is an acronym whose letter stands for some of the more common symptoms of the condition: **C**oloboma of the eye, **H**eart defects**, A**tresia of the choanae, **R**etardation of growth and development, **G**enital or urinary abnormalities, **E**ar abnormalities and deafness. CHARGE syndrome is not caused by any known exposures during pregnancy.[2] It is usually sporadic with no other affected individual in the family. Individual with CHARGE need supportive, loving homes, early intervention appropriate and challenging educational and vocational programs and preventive medical care. They also need multidisciplinary follow-up.[3]

What Genes are Related to CHARGE Syndrome?

Mutations in the CHD7 gene cause more than half of all cases of CHARGE syndrome. The CHD7 gene provides instructions for making a protein that most likely regulates gene activity (expression) by a process known as chromatin remodeling. Chromatin is the complex of DNA and protein that packages DNA in to chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression it is regulated during development.[4]

When DNA is tightly packed gene expression is lower than when DNA is loosely packed. Most mutations in the CHD7 gene lead to the production of an abnormally short and non functional CHD7 protein, which presumably disrupts chromatin remodeling and the regulation of gene expression. Changes in gene expression during embryonic development likely cause the signs and symptoms of CHARGE syndrome. Problems occur early in the first trimester, especially between the third and ninth weeks of post conception. There is a crucial stage of embryogenesis, when failure to rupture the primitive bucconasal membrane (35-38 days) brings about choanal atresia. Conotruncal cardiac defects can result from aberrations in cephalic neural crest cell migration during 4th and 5th weeks after conception. The cochlear duct begins to develop around the 36th day, and the eyes develop between days 34 and 44 days of post conception, which is also the time during which the cranial nerves are developing. All the malformations of the CHARGE syndrome occur early during the first trimester.[5, 6]

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