

THE ROLE OF GENETIC FACTORS IN CHILDREN'S HEART DISEASES

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Annotation: Heart diseases in children, especially congenital heart defects (CHD), are one of the global health problems. In recent years, the role of genetic factors in the etiology of these diseases has been increasingly studied. This article analyzes the role of genetic factors, chromosomal abnormalities, gene mutations and modern genetic diagnostic methods in the development of children's heart diseases. The role and prospects of genetic research in clinical practice are also highlighted.

Keys words: congenital heart defects, genetics, mutation, pediatrics, cardiology, genomics.

Introduction: Congenital heart diseases are among the most common birth defects in children and occur in approximately 1% of all newborns. These diseases result from disorders in the structural and functional development of the heart.

MDPI Modern scientific research shows that heart development is associated with complex genetic and molecular processes, and any genetic disorders that occur in these processes can lead to heart defects. Therefore, the study of genetic factors is one of the most relevant areas of pediatric cardiology.

Main part 1. Genetic basis of heart development: The heart is one of the first organs to develop during embryogenesis. This process occurs through a complex system involving gene expression, signaling pathways, and cell migration.

If gene expression is disrupted in this process, defects in the anatomical structure of the heart occur. For example, transcription factors such as GATA4, NKX2-5, TBX5 play an important role in heart development.

2. Genetic factors and congenital heart defects

Congenital heart diseases have a multifactorial etiology, resulting from the interaction of genetic and environmental factors.

Genetic factors are divided into the following groups:

2.1. Chromosomal abnormalities

Trisomy 21 (Down syndrome)

Turner syndrome (45,X)

According to studies, in some cases, chromosomal abnormalities are one of the important causes of CHD.

2.2. Copy Number Variation (CNV)

CNV is the gain or loss of small pieces of genetic material that affect the development of the heart. CNVs are associated with many syndromic heart diseases.

2.3. Single gene mutations

Some heart diseases are associated with specific gene mutations:

GATA6, SMAD6, FLNA

According to new research, approximately 7–10% of heart diseases in children are associated with single gene mutations.

3. De novo mutations and heredity.

Genetic studies conducted in recent years have shown that:

In many cases, mutations occur de novo (i.e., they are newly formed, not present in the parents).



In some cases, they are inherited.

De novo mutations play an important role in especially complex and syndromic heart defects.

4. Genetic diagnostic methods.

Genetic diagnostics plays an important role in modern pediatric cardiology:

4.1. Karyotyping.

Identification of chromosomal changes.

4.2. Chromosomal microarray analysis (CMA)

Effective in identifying CNVs.

4.3. Whole exome sequencing (WES).

The most modern method for identifying gene mutations.

According to studies, the rate of detection of genetic causes through the combination of CMA and WES reaches 28%.

5. Clinical significance of genetic factors.

The clinical significance of genetic diagnostics is as follows:

The possibility of early diagnosis.

Determining the prognosis.

Choosing an individual treatment strategy.

Providing genetic counseling.

Determining the genetic cause helps to assess the risk for future generations in the family.

6. Genetic and phenotypic diversity

The same gene mutation can lead to different clinical manifestations. This is explained by: Penetrance is explained by expressivity. Therefore, the clinical manifestations of genetic diseases are very diverse.

7. Future prospects.

Modern science is developing in the following areas:

Artificial intelligence-based diagnostics, gene therapy, personalized medicine.

In the future, individual treatment methods based on genetic research are expected to be widely introduced.

Conclusion: Genetic factors play an important role in the development of heart diseases in children. Chromosomal abnormalities, gene mutations and CNVs are one of the main causes of these diseases. Modern genetic diagnostic methods allow for early detection and effective treatment of these diseases. At the same time, genetic research is opening a new era in pediatric cardiology.

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