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Early Detection and Modern Treatment Methods of Congenital Heart Defect

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Annotation: This article discusses congenital heart disease, its causes, symptoms, and prevention. In addition, we will introduce modern diagnostic and treatment methods depending on the type of pathology and the general condition of the child. The issue of early detection of congenital heart disease, which is very important for improving the quality of life of the child and preventing serious complications, is raised. We will consider the genetic and environmental factors that mainly affect this disease. There are several types of congenital heart defects, depending on their anatomical location and the effect on blood flow. We will dwell on each of them below. Heart defects are the most common congenital defects and are the main cause of death in children due to developmental defects.

Keywords: congenital heart disease (CHD), atrial septal defect (ASD), ventricular septal defect (VSD), cardiac surgery, echocardiography, aortic stenosis, tetralogy of Fallot (TOF), cardiac catheterization.

Introduction: Congenital heart defects (CHD) are among the most common anomalies in newborns. According to the World Health Organization (WHO), 8-12 out of every 1000 babies are born with this disease. In some regions, this figure can even reach 15-20. Approximately 30-50% of babies born with CHD will require surgical intervention in the first year of life . If left

untreated, in severe cases, the baby may die within the first year.

Objective: The main objective of this article is to analyze the clinical and diagnostic significance of early detection of congenital heart defects (CHD), to demonstrate the effectiveness of detection using currently used modern instrumental examination methods (e.g., echocardiography, MRI, CT), and to study innovative methods of surgical and conservative treatment.

Material and methods: The article uses scientific research methods such as analysis of existing scientific literature, comparative analysis, statistical analysis, mathematical analysis. In addition, information about patients born with congenital heart defects and diagnostic methods are used.

Causes of the development of congenital heart disease:

- **Genetic factors**

The process of heart embryogenesis is a complex and genetically regulated process. Any mutations or chromosomal abnormalities that occur during this process can lead to birth defects .

Chromosomal syndromes — Down syndrome (trisomy 21), Turner syndrome, Patau and Edwards syndromes are common.

Monogenic mutations — may be associated with a mutation in a specific gene (for example, NKX2-5, GATA4 genes).

- **Infectious factors**

Certain infections that the mother has during pregnancy can cause the fetus's heart to develop abnormally.

Measles (rubella) — If contracted in the first trimester, there is a high risk of developing heart defects, vision, and hearing problems.

Cystic viruses (such as cytomegalovirus) and other TORCH-complex infections can damage heart development.

- **Toxic and pharmacological factors**

Maternal exposure to certain substances or taking medications during pregnancy can have teratogenic effects.

Alcohol — fetal alcohol syndrome is associated with heart defects.

Drugs (cocaine, amphetamine) - cause blood vessels to narrow.

- **Physical and chemical factors**

Radiation — particularly in women exposed to ionizing radiation — is at increased risk of developing a defective fetus.

Heavy metals (lead, mercury) and chemicals (pesticides) harm the development of the heart and nervous system.

- **Environmental factors**

Excessively high temperatures, especially in the first trimester, can increase the risk of heart defects (stressing the fetal cells) .

With high nitrate levels , especially in rural areas, can harm the fetus.

Substances released from industrial waste or building materials (formaldehyde, benzene, toluene) have a toxic effect on the fetus .

The most common TYUPs include:

- ✓ ventricular septal defect (VSD) - 15-42% of all TYUPs

- ✓ septal septal defect (SSD) - 6-19%,
- ✓ patent ductus arteriosus (PDA) - 5-18%,
- ✓ tetralogy of Fallot -5-14%,
- ✓ pulmonary artery stenosis - 5-9%,
- ✓ coarctation of the aorta - 4-15%,
- ✓ aortic stenosis - 3-9%,
- ✓ transposition of the great arteries - 3-6%

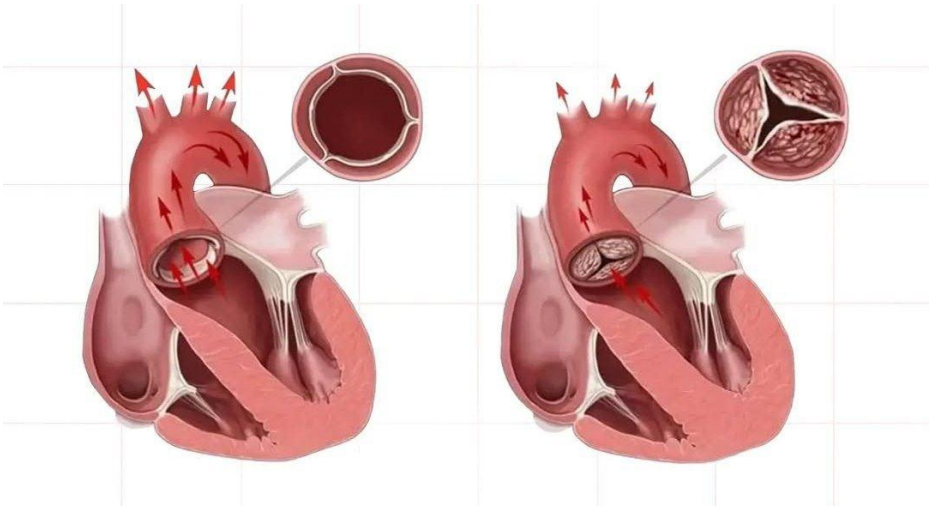

Figure 1. On the left - *a healthy heart;* on the right - *aortic stenosis type TYuP*

Gender association of congenital heart defects

congenital heart defects and gender was carried out on the basis of data collected in several large cardiac surgical centers and information from the literature. As a result of the analysis of 31,814 patients with congenital heart and great vessel defects , a clear relationship between the type of defect and the patient's gender was revealed. According to the ratio of the sexes, congenital heart defects can be divided into three groups: "male", "female" and "neutral".

Table 2: Gender ratio of patients with congenital heart and major vascular defects. *E – male, A – female*

Birth defect	Sex ratio, E:A
Patency of the ductus arteriosus	1 : 2.72
Lautembacher's disease	1 : 2.14
Secondary type of interstitial defect	1 : 1.84
Ventricular septal defect and patent ductus arteriosus	1 : 1.51
Phallus triad	1 : 1.45
Eisenmenger complex	1 : 1.40
Partial atrioventricular septal defect	1 : 1.36
Primary type of interstitial defect	1 : 1.20
Partial anomaly of pulmonary venous confluence	1 : 1.19
Ventricular septal defect	1 : 1.02
Obstructive defect between the aorta and pulmonary artery	1 : 1.01
Complete atrioventricular septal defect	1 : 1.01
Ebstein anomaly	1.02 : 1

Pulmonary artery stenosis	1.04 : 1
Tricuspid valve atresia	1.16 : 1
Common arterial trunk	1.21 : 1
Tetralogy of Fallot	1.35 : 1
Coarctation of the aorta	2.14 : 1
Aortic stenosis	2.66 ; 1

Congenital heart defects are conditionally divided into 2 groups :

1. White (arterial and venous blood do not mix, with left-right blood flow). Includes 4 groups:

- With enrichment of the small blood circulation (patent ductus arteriosus, septal defect, ventricular septal defect, AV-communicator, etc.);
- With a weakening of the small circle of blood circulation (isolated pulmonary stenosis, etc.);
- With a weakening of the large circle of blood circulation (isolated aortic stenosis, coarctation of the aorta, etc.);
- Without significant systemic hemodynamic disturbances (heart dispositions - dextro-, sinistro-, mesocardia, cardiac dystopia - cervical , thoracic, abdominal).

2. Blue (with right-left blood flow, arterial and venous blood mix). Includes 2 groups:

- With enrichment of the small blood circulation (complete transposition of the main vessels, Eisenmenger complex , etc.).
- With a weakening of the small circle of rotation (tetrad of Fallot, Ebstein anomaly, etc.)

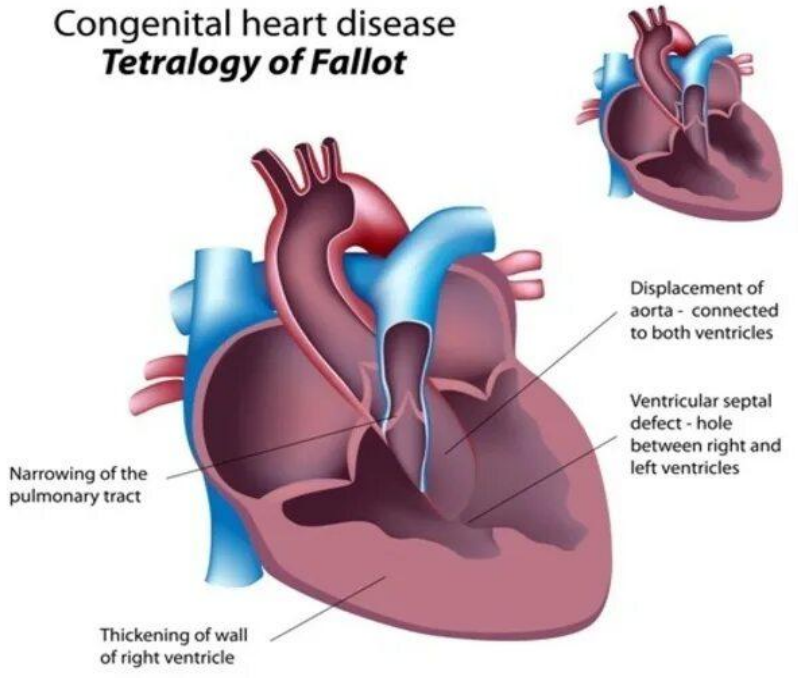

Figure 2: Blue heart disease type of congenital heart disease.

Symptoms of the disease

- ✓ Paleness of the skin and mucous membranes
- ✓ Respiratory failure
- ✓ Heart failure
- ✓ Physical development lag

- ✓ Bluish skin
- ✓ Heart murmur

Figure 3: A sign of cyanosis in congenital heart disease

Early detection methods:

1. Prenatal screening

- ➢ Ultrasound examinations during pregnancy : Used to evaluate the structure and function of the fetal heart.

2. Newborn screening:

- ➢ Pulse oximetry: In newborns, hidden heart defects can be detected by measuring blood oxygen levels.
- ➢ Physical examination : A doctor may detect heart murmurs or other abnormalities.

3. Instrumental diagnostic methods:

- ➢ Echocardiography (ExoKG): An ultrasound examination of the heart that helps detect structural and functional changes.
- ➢ Electrocardiography (ECG): To detect rhythm and conduction abnormalities by measuring the electrical activity of the heart .
- ➢ Radiography: Chest X-rays are taken to assess the condition of the heart and lungs.
- ➢ Cardiac MRI and CT: Magnetic resonance imaging and computed tomography are used to obtain detailed images of the heart.

Risk factors for having a baby with UTP:

- ✓ having children with congenital defects among close relatives,
- ✓ the pregnant woman is over 35 years old,
- ✓ endocrine diseases in the couple,
- ✓ toxicosis and risk of miscarriage in the first trimester of pregnancy,
- ✓ infectious diseases experienced during pregnancy (especially in the first trimester of pregnancy),
- ✓ history of stillbirths and miscarriages
- ✓ Smoking and alcohol consumption during pregnancy
- ✓ taking a number of medications during pregnancy.

According to the data, 29-50% of mothers with chronic alcoholism give birth to children with UTP; 3-6.2% of women with diabetes give birth to children with UTP. 5% of parents with UTP give birth to children with heart defects. Being aware of this information makes it possible to organize preventive measures during pregnancy planning and at the beginning of it.

To prevent congenital heart defects in the fetus, a pregnant woman is advised to lead a healthy lifestyle, stay in an ecologically clean area as much as possible, and avoid viral infections, especially during the critical period from 2 to 8 weeks of pregnancy.

Diagnostics

- ✓ MRI of the heart
- ✓ X-ray
- ✓ Electrocardiography (ECG)
- ✓ Echocardiography (ECHO)

Baby Diagnosis in the first year of life includes the following methods:

- ✓ Cardiac catheterization
- ✓ Aortography
- ✓ Phonocardiography

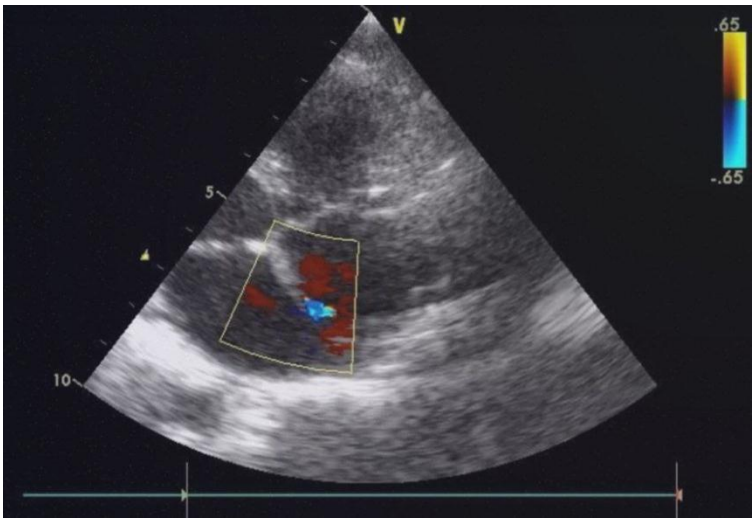

Figure 4: Early detection of congenital heart defects

Modern treatment methods

Treatment for congenital heart defects in children depends on the specific heart problem and its severity. Some congenital heart defects do not have long-term effects on the child's health. They can be safely left untreated. Serious congenital heart defects require immediate treatment once they are discovered. Treatment may include:

- ✓ Medicines
- ✓ Heart procedures
- ✓ Heart surgery
- ✓ Heart transplant

Medicines It is used with other treatments to treat complications of congenital heart defects. They help the patient increase blood pressure, reduce the workload on the heart by removing fluid from the body, and control irregular heartbeats.

Heart procedures and surgeries include:

- ✓ Cardiac catheterization. Some types of congenital heart defects in children can be corrected using thin, flexible tubes called *catheters* . Once the catheter is in place, small instruments are passed through the catheter to correct the heart condition. For example, the surgeon can repair holes or narrowings in the heart. Some catheters are done in stages over several years.
- ✓ Heart transplant. If a serious congenital defect cannot be repaired , a heart transplant may be necessary.
- ✓ with a heart murmur that is performed before birth . This procedure is rarely used and is only possible in specific situations.

High-tech surgery and innovative approaches new innovative methods are being introduced through artificial heart valves, 3D bioprinting technology , and regenerative medicine.

1. Artificial heart valves

Heart valves are important for keeping blood flowing in the right direction. Some congenital heart defects can cause heart valves to become narrowed (stenosis) or to close incorrectly (regurgitation). In such cases, the problem can be corrected by installing artificial heart valves. Currently, the type of artificial valve that is under investigation includes Growing Valves (Regenerative Heart Valves):

- ✓ New generation valves specially designed for children.
- ✓ These valves can grow with the child's heart, reducing the need for repeat surgeries.

2. Creating heart tissue using a 3D bioprinter

In recent years, *bioprinting technology* has been making great strides in medicine. This method makes it possible to create heart tissue based on living cells.

How does it work?

- ✓ Cells taken from the patient themselves are multiplied in the laboratory.
- ✓ A 3D bioprinter creates the shape of heart tissue using special biomaterials (bioink).
- ✓ The prepared tissue or valve is transplanted into the patient.

Conclusion: Congenital heart defects (CHDs) are one of the most common congenital pathologies in infants , and their early detection and modern treatment methods can significantly improve the quality of life of patients. Thanks to the development of diagnostics, including prenatal screening, pulse oximetry, echocardiography, and cardiac MRI, it has become possible to detect CHD cases at an early stage. Modern treatment approaches include advanced methods such as minimally invasive surgery, catheterization therapy , and gene therapy. Such approaches reduce the need for complex surgeries, accelerate the recovery process of patients , and help increase life expectancy.

In the future, innovations in regenerative medicine and bioengineering, such as cell therapy and the development of artificial heart tissue, will expand the possibilities for more effective treatment of congenital heart defects. Therefore, early detection of congenital heart diseases, improvement of modern treatment methods , and widespread implementation of cardiac rehabilitation remain key areas in this regard.

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