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# The Origin of Congenital Heart Defects and the Basics of Modern Clinical Diagnosis

**Yusupov Ziyodulla; Lutfullin Said; Raxmatillayev Rabbonjon**
Samarkand State Medical University Clinical Resident 1st year

***Abstract:*** Congenital heart defects are a general name for various defects in the structure of the heart and large vessels that form in a child before birth. Because of them, the heart does not function properly and blood flow in the body is impaired. This can lead to serious complications, including death. Congenital heart defects are pathological changes in the structure of the heart chambers, valves, and large vessels that lead to organ dysfunction. They are formed in the womb at 4-6 weeks of embryonic development. They can be caused by chromosomal and gene mutations, as well as the mother's lifestyle and diseases encountered during pregnancy.

***Key points:*** Congenital heart disease, Classification of heart defects, Causes of congenital heart defects, Symptoms of congenital heart defects, Complications of congenital heart defects.

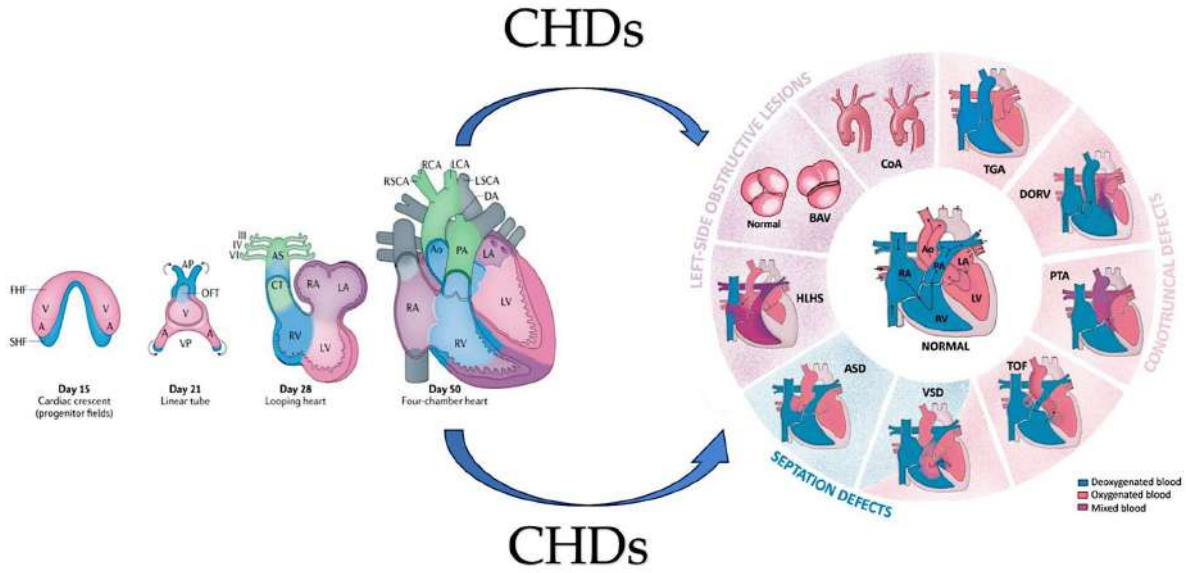
## Introduction

Such anomalies are one of the most common forms of congenital developmental disorders, occurring in 1% of children; in 20-25% of cases, congenital heart defects can lead to serious consequences and death in the first days after birth, and sometimes after a few hours, and therefore require immediate surgical intervention. Deviations in the structure of the heart and blood vessels have been and remain the main cause of infant mortality. To clearly understand how heart defects affect the functioning of the cardiovascular system, it is necessary to understand how a healthy heart works and normal blood circulation occurs. In a simplified form, this process looks like this. Dark venous blood, saturated with carbon dioxide, enters the right side of the heart through the veins from all organs and tissues. Passing through the right atrium and right ventricle of the heart, it enters the lungs through the pulmonary artery (this is the pulmonary circulation). There it is enriched with oxygen, “throws off” carbon dioxide and returns to the heart: it passes through the left atrium and ventricle, then enters the aorta (a large artery) and returns to the organs and tissues (this is the systemic circulation. ).

## Heart anatomy and blood flow direction

Normally, the left and right halves of the heart are separated from each other, the pulmonary and systemic circulation do not intersect in any way. However, with anatomical deformation of the heart, a situation may arise when a connection not provided by nature appears between them, the so-called shunt, and normal blood circulation is disrupted. Also, failure in this process can occur due to narrowing of the large vessels and malfunctioning of the valves located between the atria and ventricles and at the exit from the ventricles.

While the baby is in the womb, this does not affect his health, because he does not breathe with his lungs, but receives oxygen along with the blood through the placenta. However, after birth, anatomical anomalies of the heart and circulatory system disorders make themselves felt, which lead to disruption of the functioning of internal organs and body systems.




Currently, about 150 congenital heart defects have been described, which can be divided into several groups depending on the nature of the deformation of a particular part of the heart.

Right-to-left shunting defects. From the right side of the heart, "venous" blood enters the left, dilutes the oxygenated blood and goes with it to the tissues and organs. This group of diseases is called "blue" heart defects, because due to the circulation of darker, oxygen-depleted blood throughout the body, the lips, tongue, and nails acquire a bluish tint (cyanosis). This group primarily includes tetralogy of Fallot, which accounts for 7-10% of congenital heart defects.

Tetralogy of Fallot is a combination of four anatomical features: a septal defect between the right and left ventricles, obstruction of the vessels leaving the right ventricle by narrowing of the pulmonary valve, overdevelopment (hypertrophy) of the right ventricle, and dextraposition of the aorta, when it does not arise on the left side of the heart, as expected, but from the right.

Left-to-right shunt defects. Oxygen-rich blood that would normally leave the left side of the heart via the aorta to other organs re-enters the right side and is sent back to the lungs with venous blood. This leads to increased pressure in the pulmonary artery and overloading of the left side of the heart, which can eventually lead to heart failure. At the same time, a small amount of blood flows to the organs and tissues, as a result of which the person's skin becomes pale. Therefore, such defects are called "colorless" or white heart defects. In most cases, they develop due to defects in the septum between the ventricles and atria. This is the most common congenital heart defect (48 cases per 10,000 children).

**Cyanosis is a characteristic sign of most heart defects**

Redirected blood flow defects. Occurs when the aorta and pulmonary artery are located in an abnormal position. The aorta, which normally supplies oxygenated blood to the organs, is connected to the right ventricle instead of the left ventricle, so venous blood begins to flow through it. And oxygenated blood circulates uselessly between the lungs and the heart. This is a fatal condition that requires emergency care and surgery in the first days of the child's life.

Defects with blockage of blood flow. Narrowing of the pulmonary artery or aorta, as well as damage to the valves connecting the atria to the ventricles, causes the normal circulation of blood to cease, which can lead to the development of heart failure.

Congenital heart defects are also divided into 4 classes of severity, depending on their complexity:

I: no serious changes in cardiac anatomy, no surgical intervention required;

II: there are reversible changes in the heart parts, the heart defect has not caused other pathological changes in the body;

III: deviations in the anatomy of the heart are irreversible, but they can affect changes in the body;

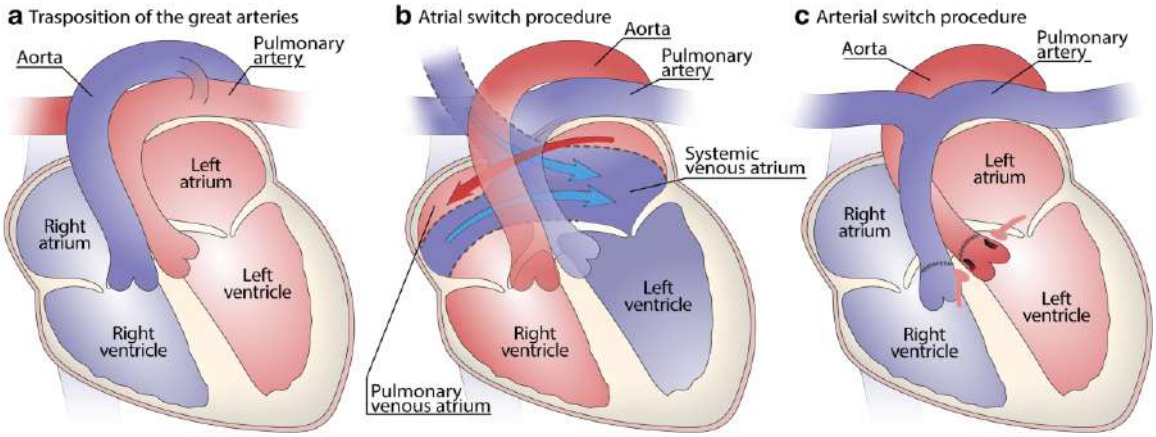
IV: changes in the heart and other organs are irreversible.

Causes of congenital heart defects

The development of birth defects is influenced by heredity and environment.

**The main reasons for the development of congenital heart defects are:**

genetic and chromosomal diseases, heredity. Up to 8% of cases of congenital heart defects are associated with chromosomal abnormalities such as Down, Edwards and Patau syndromes. Also, a congenital heart defect can be passed on to a child from one of the parents. The probability of this is from 3 to 50%;




intrauterine infections: cytomegalovirus infection, chickenpox, measles, rubella. The causative agents of these diseases can cross the placental barrier and be transmitted from mother to fetus;

chronic diseases of the mother: diabetes mellitus and other metabolic diseases, systemic lupus erythematosus;

Maternal poisoning with drugs, heavy metal salts, and toxins during pregnancy. Alcohol and drug use in early pregnancy;

exposure to ionizing radiation.

40-50% of people with Down syndrome have congenital heart defects

The lifestyle and health status of the parents at the time of conception may be additional risk factors for the development of heart defects. This anomaly is more common if the father or mother is over 35 years old, smokes, abuses alcohol, or works in hazardous jobs.

**Signs of congenital heart defects**

Congenital heart defects can present with a variety of symptoms, depending on which part of the heart is abnormal, as well as the severity of the condition and any additional abnormalities in the child's development. If the heart defect is minor, it may not manifest itself in any way for months or even years.

**Common symptoms of congenital heart defects include:**

a. delayed growth and weight gain;

b. shortness of breath during feeding, the child has difficulty sucking, takes breaks from feeding to breathe;

c. difficulty breathing;

d. high fatigue during physical activity;

e. cyanosis or, conversely, pale skin color;

f. heart murmurs;

g. frequent colds;

h. swelling;

i. anemia;

j. sleep disorder;

k. swelling and pulsation of the neck veins.

**Complications of congenital heart defects**

Serious abnormalities in the structure of the heart can lead to fatal conditions that require immediate treatment.

**Common complications of congenital heart defects include:**

a. endocarditis - inflammation of the inner lining of the heart, which disrupts the functioning of this organ and contributes to the formation of blood clots;

b. heart failure - a syndrome in which the heart begins to pump blood worse and cannot provide the necessary intensity of blood circulation;

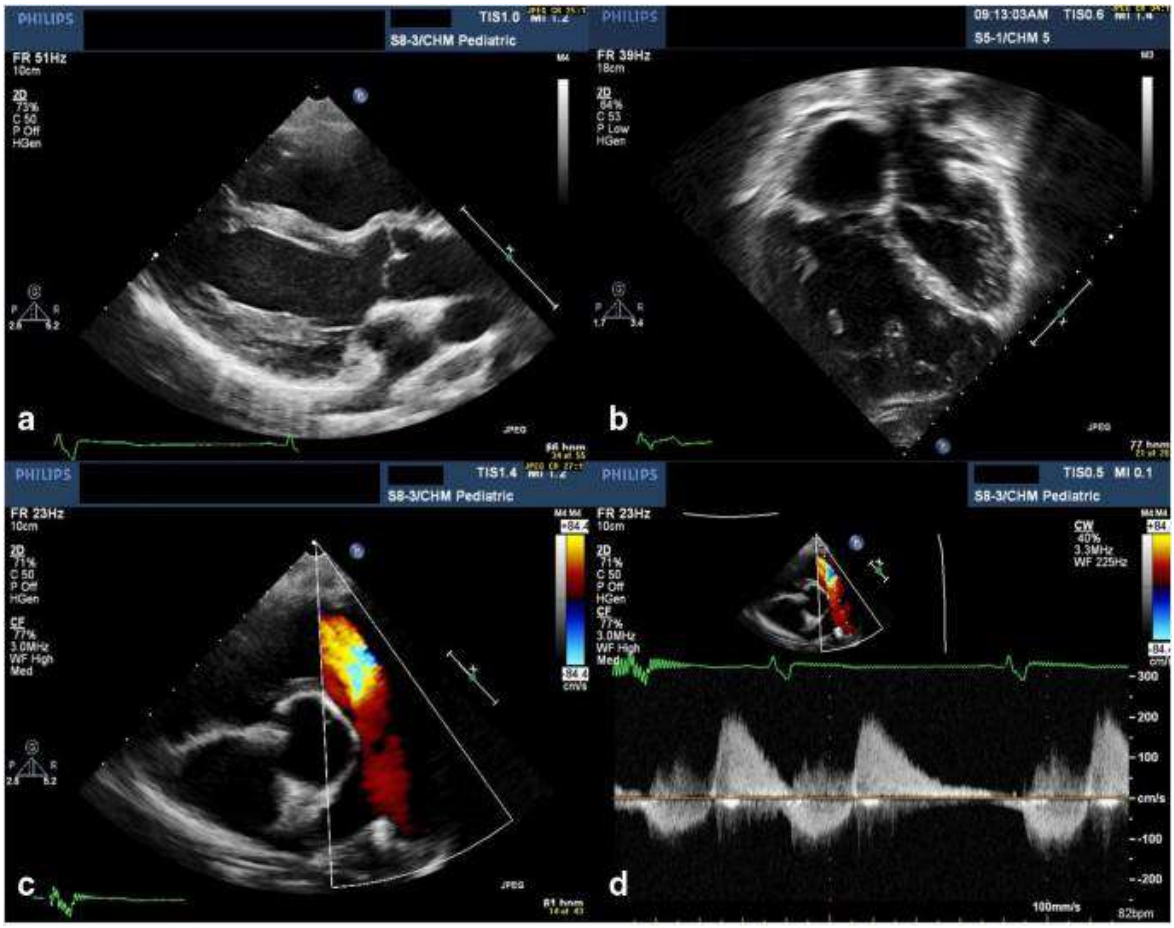
c. frequent pneumonia caused by blood stagnation in the lungs;

d. pulmonary hypertension - increased pressure in the lungs, which leads to respiratory failure;

e. angina pectoris - attacks of heart pain caused by a lack of blood supply to the organ;

f. Myocardial infarction is a fatal condition that occurs when blood flow in a coronary artery is disrupted or blocked. As a result, the heart muscle stops receiving oxygen, its cells die, and it loses its ability to contract.

Diagnosis and treatment of congenital heart defects are carried out by neonatologists, pediatricians, cardiologists and surgeons. An obstetrician-gynecologist and geneticist are responsible for prenatal diagnosis of intrauterine anomalies, which is carried out during pregnancy.



**Heart defects in a baby can be detected during pregnancy**

The most effective methods for diagnosing heart defects are instrumental. They are used both during pregnancy and after the birth of the child.

**Prenatal diagnosis**

According to clinical recommendations, every pregnant woman should undergo a screening ultrasound at 11-13 weeks of pregnancy, which can identify the risk of congenital pathologies, including heart anomalies. If, using ultrasound, the doctor detects any abnormalities that may indicate problems with the cardiovascular system, for example, an increase in the thickness of the nuchal translucency (accumulation of fluid on the back surface of the fetal body), a comprehensive examination is prescribed.

Fetal echocardiography (ultrasound examination of the fetal heart) is used for intrauterine diagnosis. A special sensor is placed on the woman's stomach, which uses ultrasound waves to create an image of the baby's heart and transmits it to a computer screen. This examination is completely safe, lasts 15-20 minutes and allows you to assess the structure and rhythm of the heart, as well as blood flow through the arteries, veins and valves.

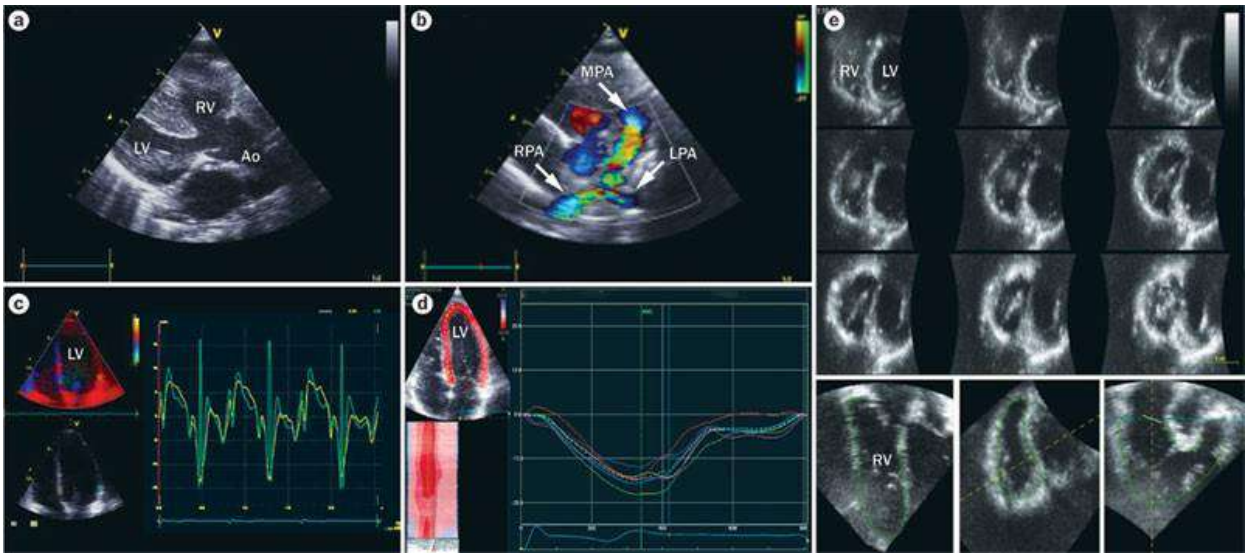
Fetal echocardiography can be performed at any stage, but this method is most informative from the 18th to the 20th week of pregnancy.

**Newborn screening**

Symptoms of heart defects in newborns are often mild, but the baby may need immediate hospital treatment or surgery. Otherwise, an undetected heart defect can lead to serious health problems or death.

Therefore, it is recommended to examine newborns with a pulse oximeter. On the first day after birth, the baby's blood oxygen saturation level is measured with a special sensor. A decrease in this indicator can indicate many (though not all) types of heart defects. In this case, a comprehensive examination of the cardiovascular system is necessary.

If oxygen saturation is below 90%, a cardiovascular evaluation should be performed.




**Other types of instrumental diagnostics**

For older children, if a heart defect is suspected, after the examination, the doctor may order additional tests to clarify the diagnosis.

The main methods of instrumental diagnosis of heart defects:

electrocardiography (ECG): allows you to detect abnormal enlargement of the heart and arrhythmias;

X-ray of the chest organs: with its help you can assess the shape and size of the heart, the state of pulmonary circulation;

phonocardiography: helps to identify heart murmurs and sounds in detail;

Echocardiography: necessary to detect defects in heart valves, septa, and blood vessels.

In severe cases, when these diagnostic methods are not enough, angiocardiography and examination of the heart chambers are performed. During angiocardiography, special tracer substances are introduced into the patient's bloodstream, and then an X-ray is taken. This method allows you to examine even the smallest vessels as accurately and precisely as possible. When examining the heart chambers, a catheter is inserted into an artery or vein, which is advanced to the heart. With the help of this study, you can assess blood flow, pressure inside the vessels, the degree of saturation of the blood with oxygen and carbon dioxide, the location, size and functions of various parts of the heart.

**Treatment of congenital heart defects**

For congenital heart defects, surgical intervention is often indicated. However, sometimes such a correction cannot correct the situation, and then a heart transplant is necessary. The operation is combined with supportive therapy. The treatment protocol in each case depends on the complexity of the defect, the patient's age and his physical condition.

Supportive therapy may include taking diuretics, which remove excess fluid from the body, reduce the load on the heart, and reduce shortness of breath and swelling; artificial ventilation; cardiotrophics (drugs that improve myocardial metabolism); symptomatic treatment.

**Prognosis and prevention**

Heart defects are one of the leading causes of death in infants and children. A quarter of such anomalies require surgical correction in the first days of life. Without surgery and supportive therapy, up to 55% of children die in the first year of life, and 85% die before the age of five. Therefore, early (primarily prenatal) diagnosis of heart defects is very important.

There is no specific prevention of cardiac abnormalities, but the risk of their development can be minimized. To do this, future parents should carefully plan their pregnancy, taking into account the family history and heredity of each. Before conception, it is necessary to abandon bad habits, undergo examinations recommended by a doctor, including tests to determine the level of immunity to infections that can be transmitted to the child in the womb and negatively affect its development.

Women in risk groups (over 35 years old, those who work in hazardous jobs, and those with heart defects) are advised to carefully monitor their health during and after pregnancy.

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