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# PREVALENCE OF CONGENITAL HEART DISEASES IN CHILDREN

**Sotvoldiyev Odiljon Komilovich**
Researcher, Andijan State Medical Institute
**Mirzakarimov Bakhromjon Xalimjonovich**
DSc of medical sciences,
Head of Department of Pediatric Surgery of
Andijan State Medical Institute

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**Abstract:**
An extensive meta-analysis of the world literature showed that congenital heart diseases were registered at a rate of 6 per 1,000 live births in 1930—1934 with its increase up to 9,1 per 1,000 after 1995. According to various estimates, the rate of congenital heart diseases was 4 to 10 per 1,000 births after 2000. However, the true prevalence of the above defects may be much higher. Most authors are in agreement that the rate of congenital heart disease varies between 19 to 75 per 1,000 live births; serious anomalies being registered at a rate of 19,1—23,9 per 1,000 births. Many cardiologists do not consider mild defects to be diseases; however, this statement calls for further investigation. Fetal echocardiography and genetic studies are of great importance in estimating the prevalence of congenital heart disease. Tables of chromosomal and monogenic syndromes associated with cardiac malformations are given.

**Keywords:** Children, congenital heart diseases, prevalence, fetal echocardiography, prenatal diagnosis, defect severity, genetic syndromes

Congenital heart defects account for 1/3 of all congenital malformations and are one of the main causes of infant mortality. In an extensive meta-analysis of world literature data, congenital heart defects were recorded with a frequency of 6 per 1000 live births in 1930-1934. with an increase to 9.1 per 1000 after 1995. According to various estimates, the incidence of congenital heart defects after 2000 ranged from 4 to 10 per 1000 children born. The true prevalence of heart defects may be much higher. According to indicators from 62 registers, their frequency can reach 50 per 1000 live births.

The congenital defect can be recognized antenatally by ultrasound. The ultrasound screening examination scheme in Russia is regulated by Order No. 808n of the Ministry of Health of the Russian Federation dated October 2, 2009. Initially, a heart defect may be suspected when the thickness of the fetal nuchal space increases during the first antenatal screening at 12-14 weeks of pregnancy, when chromosomal abnormalities are excluded (or detected) and the most severe structural defects of organs can be detected. If the thickness of the fetal nuchal space is ≥3.5 mm, then the risk of congenital heart disease is 4 times higher than in fetuses without this sign [4]. The diagnosis of congenital heart disease in the fetus is clarified at 17-22 weeks of pregnancy, since many heart defects are recognized later than the first ultrasound screening. The use of color Doppler mapping in this case is extremely important, since it allows one to determine defects of the great vessels.

Prenatal ultrasound diagnostics is one of the important factors in reducing infant mortality from congenital heart defects, however, the resolution of the method is closely related to the level of the hospital and the qualifications of the doctor performing fetal echocardiography. The average detection rate of heart defects in Europe with a single ultrasound scan is 46%, with a repeat scan - 56%, with repeat scans in specialized centers - 95%. According to the pediatric cardiology department of the San Francisco Medical University Hospital, among infants with hypoplastic left heart syndrome (n=81) admitted during the period 1999-2010, 49 (60.5%) were diagnosed antenatally, and postnatally - in 32. With the widespread use of fetal echocardiography, the number of cases of detection of heart defects during fetal development has significantly increased. This is especially true for children with so-called "ductus" dependent defects. If such newborns previously died in the first hours or days of life, then by establishing a diagnosis in utero, cardiac surgeons can prevent death.

Among infants with heart defects, the proportion of premature infants is 2 times higher than in the general population of newborns. The risk of congenital heart defects is increased for preterm infants who are born as a result of spontaneous preterm birth. The risk is high specifically for heart abnormalities (chromosomal diseases and multiple malformations are excluded from the group). This is probably due to the effect of teratogens, among which congenital infections play an

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important role, which are one of the main causes of premature birth.

Division of congenital heart defects depending on the severity of the defect according to J.I. Hoffman the following:

A. Heavy

I. Cyanotic heart defects

1. d-transposition of the great vessels.
2. Tetralogy of Fallot, including pulmonary atresia or absence of the pulmonary valve.
3. Hypoplasia of the right ventricle:

a) atresia of the tricuspid valve;

b) pulmonary atresia with an intact interventricular septum;

c) Ebstein's anomaly.

4. Hypoplasia of the left ventricle;

a) aortic atresia;

b) mitral valve atresia.

5. Single ventricle.
6. Double origin of the great vessels from the right ventricle.
7. Common arterial trunk.
8. Total anomalous drainage of the pulmonary veins.
9. Critical pulmonary artery stenosis.
10. Combined atypical anomalies such as double origin of the great vessels from the left ventricle, some forms of L-transposition of the great arteries.

II. Acyanotic defects

11. Atrioventricular communication.
12. Large ventricular septal defect.
13. Large patent ductus arteriosus.
14. Critical or severe aortic stenosis.
15. Severe pulmonary stenosis.
16. Critical coarctation of the aorta.

B. Moderate severity

1. Moderate or severe aortic stenosis or aortic insufficiency.
2. Severe pulmonary artery stenosis or insufficiency.
3. Non-critical coarctation of the aorta.
4. Large atrial septal defect.
5. Complex form of ventricular septal defect.

C. Mild

1. Small defect of the interventricular septum.
2. Small patent ductus arteriosus.
3. Mild pulmonary stenosis.
4. Bicuspid aortic valve without stenosis or insufficiency.
5. Small or spontaneously closing atrial septal defect.

The number of registered atrial septal defects does not include a patent foramen ovale (a defect in the area of the oval fossa up to 0.2 cm in size), which occurs in 15-30% of adults, is not accompanied by hemodynamic disorders, for this reason does not require treatment and does not affect the duration life. For example, the bicuspid aortic valve, the most common defect, is generally excluded from this assessment. The indicated number of defects does not include minor defects of the ventricular septum, patent ductus arteriosus in premature infants, bicuspid aortic valve, or patent foramen ovale. If children with a bicuspid aortic valve are included, the number of congenital heart defects reaches 20.1 per 1000 live births, and when all minor defects are included (atrial septal aneurysm, isolated lobar pulmonary vein anomalies, small asymptomatic patent ductus arteriosus, very small ventricular septal defect) - 75 per 1000.

Bicuspid aortic valve is associated with significant morbidity and mortality later in life, with an incidence of 10 to 20 per 1000 in the general population. Recent studies have established a high degree of heritability of bicuspid aortic valve alone or with other cardiovascular anomalies, especially in combination with left ventricular outflow tract obstruction.

If isolated atrial septal aneurysm and persistent left superior vena cava are taken into account (each defect occurs in 5-10 per 1000 newborns), then the incidence of heart defects approaches 75 per 1000 newborns. In light of the above, the indicated frequency is quite realistic.

In 2000, the total number of newborns worldwide with congenital heart defects was approximately 623,000 (320,000 with simple defects, 165,000 with moderately complex defects, and 138,000 with complex heart defects).

Indicative are the results of the EUROCAT study (European Surveillance of Congenital Anomalies), which included data from registers of all congenital malformations in newborns in European countries (1.5 million births annually from 22 countries) for 2003-2007. According to this study, serious congenital anomalies malformations were recorded with a frequency of 23.9 per 1000 births, of which 80% of children were born alive, 2.5% were born with congenital anomalies and died in the 1st week of life, 2.0% were cases of stillbirth or fetal death after 20 weeks of gestation, 17.6% - termination of pregnancy during prenatal diagnosis of a congenital malformation. Most children born with heart defects do not have other birth defects; in 25-40% of cases, heart defects occur in association with other anomalies or as part of a syndrome. In addition, about 30% of children with chromosomal abnormalities have congenital heart disease. A significant percentage of heart defects are associated with aneuploidy or changes in the number of chromosomes, as well as with monogenic genetic syndromes. These data indicate that, despite the multifactorial genesis of most congenital heart defects

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and the role of unfavorable factors in the antenatal period, the genetic component has a very significant influence on the prevalence of defects. In practice, it is often not possible to perform molecular genetic typing for monogenically inherited heart defects, and the genetic defect can only be determined in a small number of children.

Congenital heart defects in adults

Enormous advances in medical and surgical care for children with heart defects over the past decade have made it possible to increase their life expectancy. Researchers estimate that in 2000, there were a total of 787,000 adults worldwide living with congenital heart disease (368,800 with simple heart disease, 302,500 with moderately complex heart disease, and 117,000 with very complex heart disease). This estimate of the prevalence of the defect in the adult population is likely to be an underestimate, since many adult patients were lost to follow-up. It has been estimated that the global population of adults with congenital heart defects is growing at approximately 5% per year and exceeded 1 million in 2005. This means that the number of such adults has exceeded the number of children with congenital heart defects. In this regard, it is extremely important to provide appropriate care and treatment for this growing population of patients, the management of which only pediatricians and pediatric cardiologists are familiar with

**CONCLUSION.** Thus, most authors agree that the incidence of congenital heart defects varies from 19 to 75 per 1000 live births, with major congenital defects reported at a rate of 19.1 to 23.9 per 1000 births. Many cardiologists do not consider mild heart defects to be a defect, but this situation needs further study. Fetal echocardiography and genetic research methods are of great importance in diagnosing and determining the prevalence of heart defects. At the same time, it is necessary to develop a special monitoring system for such patients, which could provide adequate control over their condition, especially taking into account the specific nature of the problems of postoperative management after complex hemodynamic palliative correction procedures over many years, not only in childhood, but also in older age.

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