ISSN: 2692-5206
AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

# CONGENITAL HEART DEFECTS IN CHILDREN (LITERATURE REVIEW)

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Congenital heart defects (CHDs) represent one of the most significant challenges in pediatric cardiology and cardiovascular surgery worldwide [1]. According to the World Health Organization (WHO), the incidence of CHDs ranges from 8 to 12 per 1,000 live births [2]. In the structure of childhood mortality, cardiovascular pathology remains one of the leading causes, with CHDs accounting for 30–45% of all mortality associated with congenital developmental anomalies [3].

The formation of the heart in the fetus occurs during the earliest stages of embryogenesis, when the basic structures of future organs and systems are being laid down [5]. External and internal factors, such as genetic mutations, infectious agents, toxic exposures, and metabolic disorders in the mother, can influence the process of heart and major vessel formation, leading to various developmental defects [6]. Additionally, several studies highlight the significant role of chromosomal abnormalities and syndromes in which the incidence of CHDs is significantly higher than in the general population (e.g., Down syndrome or DiGeorge syndrome) [7].

Modern methods of prenatal diagnosis, particularly fetal echocardiography, allow for the detection of some defects as early as 18–20 weeks of gestation [8]. However, not all changes in the cardiovascular system can be visualized prenatally, especially in cases of minor defects or defects with mixed hemodynamics [9]. Timely diagnosis is critical for determining the management strategy for the newborn (e.g., the need for emergency surgical intervention or transfer to a specialized hospital) and reducing the risk of adverse outcomes [10].

According to leading Russian clinics, there has been a relative decrease in severe forms of CHDs in recent years due to the wider use of early ultrasound screening during pregnancy [4]. However, some authors emphasize that in many regions, the level of specialized care for expectant mothers remains insufficient, access to high-precision diagnostic equipment is limited, and extended screening for chromosomal and genetic abnormalities is not routinely performed [11]. As a result, children with CHDs do not always receive timely and accurate diagnosis and treatment, which negatively affects survival rates and quality of life [12].

Significant interest lies in research on the structural classification of congenital heart defects, which reflects the complexity and variability of defects in both morphological and hemodynamic terms [13]. The most common forms include ventricular septal defects (VSD), patent ductus arteriosus (PDA), atrial septal defects (ASD), tetralogy of Fallot, transposition

ISSN: 2692-5206
AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

of the great arteries (TGA), coarctation of the aorta, and others [14]. Critical CHDs requiring urgent surgical intervention in the neonatal period (e.g., hypoplastic left heart syndrome) are less common but have extremely high mortality rates in the absence of timely surgery [15].

According to several Russian authors, risk factors for CHDs include an unfavorable obstetric and gynecological history (infections during pregnancy, use of teratogenic medications), maternal age (over 35 years), environmental factors (air pollution, radiation), and hereditary predisposition [16]. The polyetiological nature of CHDs is emphasized: often, multiple causal factors may contribute to the formation of a single defect [17].

In pediatric practice, clinical symptoms play a crucial role, directly depending on the type and severity of the defect and the degree of hemodynamic impairment [18]. In some children, the defect may manifest immediately after birth (cyanosis, dyspnea, heart failure), while in others, symptoms may appear later in childhood (episodes of weakness, fatigue, delayed physical development) [19]. Diagnosis of CHDs, in addition to standard methods (physical examination, electrocardiography, chest X-ray), includes echocardiography, Doppler ultrasound, and often cardiac catheterization and angiocardiography [20]. Modern treatment approaches are based on interventional cardiology and cardiac surgery, with the timing and method of intervention depending on the morphology of the defect and the child's overall condition [21].

One of the most pressing issues remains the determination of prognostic factors for outcomes, particularly long-term survival, quality of life, and physical and psychomotor development in children with CHDs [22]. Several large multicenter studies show that with timely and adequate surgical treatment, many children can achieve satisfactory or even normal health levels [23]. However, the presence of severe associated developmental defects (including those outside the cardiovascular system), genetic syndromes, and other complicating factors can significantly worsen the prognosis [24].

Thus, the analysis of scientific literature indicates the high prevalence of congenital heart defects and their significant impact on childhood morbidity and mortality rates. Despite advances in modern diagnosis and treatment, challenges remain in early detection, preoperative preparation, and rehabilitation of children with CHDs, as well as in prevention and genetic counseling [25]. This article provides a literature review aimed at systematizing current knowledge on congenital heart defects in children, from epidemiology and classification to diagnostic and therapeutic approaches.

**Brief Overview of Epidemiology and Etiology of CHDs**

Congenital heart defects account for a significant proportion of all congenital anomalies: according to various authors, up to 30% of children with any congenital anomalies have cardiovascular system pathologies [1]. Overall, the global prevalence of CHDs is estimated at 0.8% to 1.2% per 1,000 live births, although these rates may vary in different regions [2,3].

Risk factors for CHDs can be divided into genetic, intrauterine (antenatal), and external (environmental, socio-domestic) factors. The genetic nature of CHDs is supported by the presence of chromosomal abnormalities (e.g., trisomy 21, 13, 18) and microdeletions

ISSN: 2692-5206
AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

(22q11.2—DiGeorge syndrome), which significantly increase the risk of heart defects [7]. There are also several monogenic mutations that affect the process of cardiogenesis [6].

Intrauterine factors may include maternal infections during pregnancy (rubella, cytomegalovirus, toxoplasmosis), maternal diabetes, hormonal dysfunctions, toxic exposures (alcohol, smoking, drugs), and the use of certain pharmacological agents with teratogenic potential [16]. The literature also highlights the role of chronic stress and adverse environmental factors (air pollution, high doses of radiation) [11]. Maternal age over 35 years, especially with a family history of CHDs, is also an important predisposing factor [7,17].

**Classification of Congenital Heart Defects**

Various classification systems for CHDs are proposed in domestic and foreign literature, but the most common approach is to categorize defects based on hemodynamic characteristics, such as defects with increased pulmonary blood flow, decreased pulmonary blood flow, combined defects, and "cyanotic" and "acyanotic" defects (based on the presence or absence of cyanosis) [18].

Defects with increased pulmonary blood flow include, for example, ventricular septal defects (VSD), atrial septal defects (ASD), patent ductus arteriosus (PDA), and other anomalies associated with left-to-right shunting [26]. "Cyanotic" defects (e.g., tetralogy of Fallot, transposition of the great arteries) are characterized by cyanosis due to the shunting of venous blood into the systemic circulation [15]. There are also complex combined defects involving multiple components (e.g., triads or pentads of Fallot).

Some authors distinguish critical CHDs, where surgical intervention or emergency palliative measures are necessary for the child's survival (e.g., hypoplastic left heart syndrome, total anomalous pulmonary venous connection) [21]. This classification has practical significance, as it determines the urgency of surgical treatment and the need for specialized resuscitation measures in the delivery room or during the first days of life [27].

**Clinical Manifestations**

The clinical manifestations of congenital heart defects in children vary depending on the type of defect, the severity of hemodynamic disturbances, and the degree of compensation [19]. The most severe forms typically manifest immediately after birth or in the first weeks of life (severe cyanosis, respiratory failure, circulatory disturbances), while milder defects (e.g., small VSD) may remain asymptomatic for a long time [9,18].

Key symptoms that may suggest CHDs in neonates include:

- Severe dyspnea or cyanotic episodes;

- Cyanosis of the nasolabial triangle, mucous membranes, or skin (especially during crying or feeding);

- Tachycardia or, conversely, bradycardia;

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AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

- Signs of heart failure (hepatomegaly, edema, poor weight gain);

- Characteristic auscultatory findings (murmurs, accentuated second heart sound over the pulmonary artery, etc.) [19,20].

In older children, if the defect is non-critical and did not manifest in the neonatal period, symptoms may include fatigue, dyspnea during physical activity, frequent respiratory infections, and delayed physical development [8,22].

**Modern Diagnostic Methods**

Standard diagnostic methods for CHDs in pediatric practice include:

1. Clinical examination: history taking (including family history), physical examination, auscultation, and assessment of skin and mucous membrane color [19].

2. Electrocardiography (ECG): allows for the evaluation of rhythm, conduction, and cardiac chamber hypertrophy [20].

3. Chest X-ray: provides information on heart size and pulmonary vascular markings [5].

4. Echocardiography with Doppler: the primary method for visualizing cardiac anatomy and major vessels, detecting septal defects, abnormal blood flows, valve regurgitation, etc. [8,28].

5. Fetal echocardiography: performed prenatally (from 18–20 weeks of gestation) to detect major CHDs and assess fetal blood flow [10].

6. Cardiac catheterization and angiocardiography: invasive methods used in complex diagnostic cases and for clarifying hemodynamic parameters before surgical intervention [20].

7. Genetic testing: indicated in cases of suspected chromosomal abnormalities or hereditary syndromes associated with CHDs [29].

In recent years, advanced imaging techniques such as cardiac magnetic resonance imaging (MRI) and multislice computed tomography (CT) with three-dimensional reconstruction have gained wider use [30]. These methods provide detailed visualization of the defect's structure and aid in planning surgical interventions.

**Medical Therapy and Palliative Measures**

In most cases, congenital heart defects with significant hemodynamic disturbances require surgical correction. However, medical therapy is often used in preparation for surgery and to maintain compensation [31]. Medications of choice include diuretics (furosemide, spironolactone), inotropic agents (cardiac glycosides), vasodilators (ACE inhibitors), beta-blockers, and others [32]. In some defects (e.g., PDA) in preterm infants, ibuprofen or indomethacin may be used to close the ductus arteriosus [33].

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Surgical Treatment

The primary method for radical correction or repair of CHDs is surgical intervention. Depending on the type of defect, the child's age, and condition, surgeries may be performed on an emergency, early, or delayed basis [14,21]. The main types of interventions include:

- Radical: completely correcting the anatomical defect (e.g., VSD closure, ASD repair, PDA ligation).

- Palliative: improving hemodynamics without fully correcting the defect (e.g., aortopulmonary anastomosis, shunt creation).

- Single-stage and multi-stage (staged correction): particularly relevant for complex combined defects (tetralogy of Fallot, transposition of the great arteries, hypoplastic left heart syndrome) [27].

One of the achievements of modern cardiac surgery is the widespread use of endovascular (catheter-based) interventions: occluder placement for ASD and VSD, endovascular closure of PDA, and stenting of narrowed segments (e.g., in coarctation of the aorta) [34]. These methods are less invasive, reduce hospitalization time, and are highly effective for certain indications.

Rehabilitation and Long-Term Outcomes

Rehabilitation of children who have undergone surgical treatment for CHDs includes a range of measures: medical support, hemodynamic monitoring, nutritional and physical activity adjustments, and psychological and educational assistance [35]. Regular follow-up with a cardiologist and pediatrician allows for the timely detection of potential complications (restenosis, arrhythmias, heart failure) and adjustment of therapy [36].

The prognosis for children with CHDs has significantly improved compared to 20–30 years ago. According to some data, with timely radical surgery, children with the most common defects (VSD, ASD, PDA) can achieve nearly normal life expectancy [23]. However, for critical CHDs requiring multi-stage interventions, the risk of complications and mortality remains high [15,27].

Prevention of Congenital Heart Defects

Prevention of CHDs aims to reduce the risk of defects during the intrauterine period and to ensure early detection and timely treatment [17,25]. Prenatal counseling for expectant parents, considering genetic factors, and maintaining a healthy lifestyle during pregnancy (avoiding alcohol and other teratogenic substances, managing chronic conditions) play a crucial role [7,16].

In cases of a family history of CHDs or suspected genetic disorders, genetic screening and high-precision prenatal diagnosis (non-invasive prenatal testing, amniocentesis if necessary) are recommended [29]. Folic acid supplementation during pregnancy planning is also

ISSN: 2692-5206
AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

advised, as it not only reduces the risk of neural tube defects but may also positively impact overall fetal development [37].

Timely ultrasound screening during the first and second trimesters of pregnancy (including fetal echocardiography) allows for the detection of most severe forms of CHDs and determines the management strategy for delivery (e.g., delivery in a specialized perinatal center with the capability for neonatal surgery within the first hours of life) [10,38].

**Discussion**

The analysis of modern scientific literature highlights the significant impact of congenital heart defects as one of the leading causes of childhood mortality and disability [3,4]. Advances in medical science and technological progress in perinatal diagnosis and cardiac surgery have led to substantial improvements: survival rates for severe CHDs have increased, and early mortality rates have decreased [15,23].

However, according to some authors, challenges remain in the uneven availability of high-tech care across regions and insufficient prenatal screening coverage [11,25]. Additionally, organizational issues related to the creation of multidisciplinary teams (obstetricians, cardiologists, neonatologists, cardiac surgeons) working in close collaboration for timely diagnosis and treatment of CHDs are crucial [39].

Further research is needed to clarify the genetic mechanisms underlying CHDs, which could lead to the development of primary prevention methods. Currently, molecular genetic research aimed at identifying specific genes responsible for cardiogenesis and mutations that increase the risk of CHDs is actively progressing [6,7].

Long-term follow-up of children with CHDs, including postoperative outcomes, will provide a better understanding of factors influencing quality and length of life in these patients. Research on psychosocial aspects is also important, as children with severe defects may experience psychological difficulties and physical limitations, affecting their socialization [35,40].

**Conclusion**

1. Congenital heart defects in children remain a pressing issue in pediatrics and pediatric cardiac surgery, contributing significantly to childhood mortality and disability.

2. The main risk factors include genetic and chromosomal abnormalities, adverse pregnancy conditions, teratogenic exposures, and environmental factors.

3. Modern diagnostic methods (prenatal echocardiography, echocardiography with color Doppler, CT, and MRI of the heart) enable early detection of CHDs and planning of timely surgical or interventional interventions.

4. Surgical and endovascular interventions allow for the radical correction of many forms of CHDs; however, long-term rehabilitation and follow-up are necessary.

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AMERICAN ACADEMIC PUBLISHER
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5. Prevention of congenital heart defects involves a range of measures—from pregnancy planning and proper prenatal care to promoting healthy lifestyles and genetic counseling.

The development of a multidisciplinary approach, further advancements in diagnostic and surgical technologies, and research into the genetic basis of CHDs offer opportunities for reducing the incidence of CHDs and improving the quality of life for affected children.

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