

**MODERN PRINCIPLES OF CONGENITAL
HEART DEFECTS IN CHILDREN***Gulzhazira Sakenovna Zhaulibayeva**Laylo Valizhanovna Kadirova**<https://orcid.org/0009-0009-0011-9363>**E-mail: kadirova.laylo@bsmi.uz**Bukhara State Medical Institute*

Abstract: This article examines the main factors of congenital heart defects: their primary causes, common types, and impact on cardiac and systemic hemodynamics. It also discusses the role of folic acid in prevention and UN goals for reducing child mortality from these conditions.

Keywords: congenital heart defects, etiology, cardiac and systemic hemodynamics, phases, genetic and environmental factors.

Резюме: В статье рассмотрены основные факторы врождённых пороков сердца: их основные причины, распространённые виды, влияние на сердечную и системную гемодинамику. Также рассмотрены значения фолиевой кислоты в профилактике и цели ООН по снижению детской смертности от этих состояний.

Ключевые слова: врождённые пороки сердца, этиология, сердечная и системная гемодинамика, фазы, генетические и экологические факторы.

Relevance. In 1971, the definition of congenital heart defects was formulated. According to S. C. Mitchell, "CHD is a structural anomaly of the heart or intrathoracic great vessels that actually or potentially results in functional impairment" [8]. The heart begins to form in the early stages of embryonic development. During gastrulation (between days 7 and 17), cardiogenic cells of the mesoderm appear on both sides of the midline, forming the first and second cardiac fields. They are located in the center of the ventral region of the embryo and form the cardiac ridges. The cells of the first cardiac field give rise to the left ventricle, left and right atria. The cells of the second cardiac field are the precursors of the right ventricle, some parts of the atria and the outflow tract [15]. Anomalies in the anatomical development of the heart and large vessels usually develop between the 2nd and 8th weeks of intrauterine development as a result of disruption of embryonic morphogenesis. They can be caused by both hereditary (genetic, chromosomal, genomic, zygotic mutations) and environmental factors affecting the developing fetus [5].

CHD can be caused by genetic or environmental factors, often a combination. Genetic mutations are caused by three main mutagens: a) physical mutagens—ionizing

radiation; b) chemical mutagens—varnishes, dyes, phenols, nitrates, benzopyrene from tobacco smoking, alcohol consumption, hydantoin, lithium, thalidomide, teratogenic drugs—antibiotics and antiviral drugs, NYQP, etc.); c) Biological mutagens—the presence of rubella virus in the mother's body, which causes congenital rubella and Gregg's triad; systemic lupus erythematosus; diabetes mellitus; and phenylketonuria. Uncorrected phenylketonuria in women of reproductive age has a six-fold higher risk of having a child with congenital heart disease [9, 12]. Congenital heart disease may be associated with a genetic syndrome. Many children with an extra chromosome 21 have heart defects. A missing fragment of genetic material on chromosome 22 causes heart defects [1].

A. Miller et al. found an increased risk of severe congenital heart disease associated with maternal age over 35 years. For conotruncal defects, the odds ratio was 1.3, for transposition of the great vessels - 1.7, for coarctation of the aorta - 1.54, for VSD - 1.20, for ASD - 1.36[14].

Main factors:

1. Impaired cardiac hemodynamics → stress on the heart chambers due to volume (valvular insufficiency and septal defects) or resistance (stenosis of vessels or orifices) → exhaustion of the compensatory mechanisms involved (Anrep's homeometry to resistance and Frank-Starling heterometry to volume) → development of hypertrophy and dilation of the heart chambers → development of heart failure (and, accordingly, impaired systemic hemodynamics).

2. Impaired systemic hemodynamics (anemia of pulmonary circulatory integrity, anemia of systemic circulation) development of systemic hypoxia (mainly circulatory in white lungs, hemic in blue lungs, as well as ventilatory and diffuse hypoxia in acute left ventricular hypoxia) [9].

Three phases are defined according to the course of congenital heart disease: a) primary adaptation - after birth, the child's body adapts to the hemodynamic disturbances caused by congenital heart disease. Insufficient compensation capabilities and the unstable condition of the child at an early age sometimes lead to a severe course of the disease and even a fatal outcome; b) relative compensation - occurs in the 2nd-3rd year of life and can last for several years. The child's condition and development improve due to hypertrophy and hyperfunction of the myocardium of different parts of the heart; c) terminal (irreversible) - it is associated with gradually developing myocardial dystrophy, cardiosclerosis, and decreased coronary blood flow [2].

The A.N. Bakulev Institute of Cardiovascular Surgery of the USSR Academy of Medical Sciences developed a classification based on the distribution of congenital heart defects, taking into account their anatomical features and hemodynamic disturbances. Practicing cardiologists find it more convenient to use a more simplified division of congenital heart defects into three groups: a) a pale type of congenital heart

defect with arteriovenous shunt – ventricular septal defect, atrial septal defect, patent ductus arteriosus; atrioventricular communication; b) a cyanotic type of congenital heart defect with venoarterial shunt – transposition of the great vessels, tetralogy of Fallot, triad of Fallot, tricuspid atresia, etc.; c) congenital heart defects without shunting, but with an obstruction to blood flow from the ventricles – pulmonary artery and aortic stenosis, coarctation of the aorta [4].

The incidence of multiple developmental anomalies in children with congenital heart defects has been studied in a number of studies. Thus, according to the data of A. Miller et al., in 71% of cases, congenital heart defects were isolated, in 13.5% there were multiple congenital malformations, and in 13%, an association of congenital heart defects with genetic syndromes was found. The most common congenital anomalies with multiple congenital anomalies were ASD (18.5%) and conotruncal defects (16%). The most common extracardiac disorders with multiple congenital malformations were anomalies of the musculoskeletal system (up to 35%), gastrointestinal tract (25%), and genitourinary system (23%). Among congenital heart defects associated with genetic syndromes, the atrioventricular canal was most common (67%) [13]. According to the study by Khruska A., Kainap S. and Rachitsan A.L., published in the journal "Bioflux" (No. 5), patients with congenital heart disease often have combinations with anomalies of the musculoskeletal system (15.5%), craniofacial dimorphism (8%), gastrointestinal tract (5%), urinary tract (4%), hemangiomas and eye pathologies (3%) and lung defects (2%) [10].

Common types of congenital heart disease:

- ventricular septal defect (VSD) - 15-23%;
- transposition of the great vessels (TGV) - 9-20%;
- tetralogy of Fallot (TOF) - 8-14%;
- coarctation of the aorta - 6-15%;
- patent ductus arteriosus (PDA) - 6-18%;
- patent atrioventricular canal - 2-5%;
- atrial septal defect (ASD) - 2.5-16%;
- anomalous pulmonary venous drainage - 0.5-2%;
- aortic stenosis - 2-7%;
- pulmonary artery stenosis - 6.8-9% [11].

A study by C.J. Alverson et al. found an association between maternal smoking in the first trimester of pregnancy and a 1.4-fold increase in the risk of atrial septal defect, 1.3-fold increase in right ventricular anomalies, 1.4-fold increase in pulmonary artery stenosis, 1.9-fold increase in common aortic trunk, and 1.8-fold increase in transposition of the great vessels [3].

It has now been proven that folate deficiency in the first trimester leads to impaired cardiac development. Taking folic acid during pregnancy is an effective

preventative measure for CHD, reducing the risk by 28-39% [7].

In 2015, the UN set a goal to reduce child mortality from congenital heart defects (CHD) by one-third by 2030. Over the past 30 years, developed countries have made significant progress in the treatment of CHD thanks to advances in interventional cardiology and surgery. However, due to high rates in developing countries, the overall global mortality rate from CHD remains high. Despite this, positive trends are observed: while in 1990 the mortality rate was 7.1 cases per 100,000 children, by 2019 this rate had decreased to 2.8 per 100,000 [6].

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