Epidemiology and risk factors for congenital heart defects in children

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Abstract: Congenital heart defects are an important problem in pediatrics due to their high prevalence and the need for early surgical correction due to significant health problems and disability in children. According to WHO, congenital heart disease occurs in 0.7-1.7% of newborns. In recent decades, the structure of cardiovascular pathology in childhood has changed significantly. Against the background of an increase in the number of cases of CHD, cardiac arrhythmias and conduction disorders, cardiomyopathies, the proportion of rheumatic diseases and infectious lesions of the myocardium decreased. CHD remains one of the leading causes of infant mortality, ranking second. In the perinatal period, congenital heart diseases cause 2.5% of deaths (0.25 cases per 1000 births), in the first year of life - 6-11% of deaths and about 50% of deaths associated with congenital malformations. In this regard, the issue of studying the epidemiology and risk factors for the development of congenital heart defects at an early age remains relevant and requires scientific research in this direction.

Keywords: congenital heart disease, epidemiology, risk factors, intrauterine infection

The birth rate of children with congenital heart disease and large vessels in different countries of the world has a very wide range of fluctuations, and in the regions of the Russian Federation, according to official statistics, it is 3.17-8.0 per 1000 newborns. Hoffman J.I.E., Kaplan S. provide data on the prevalence of congenital heart disease in the USA, which fluctuates from 4 to 50 per 1000 newborns, while the frequency of moderate and severe forms of congenital heart disease is on average 6%.

In Uzbekistan, the birth rate of children with congenital heart defects ranges from 5.5 to 15.7 people per 1000 live births.

To date, more than 90 anatomical variants of congenital heart defects and about 200 different combinations of them have been described in domestic and foreign literature. In different regions of the country, heart defects and large vessel defects occupy 1-2 places in the structure of all congenital heart defects, competing for first

place with defects of the musculoskeletal system, in particular, with developmental anomalies of the musculoskeletal system.

The frequency of congenital heart disease depends on the development of the health care system in the country or the diagnostic methods available to the doctor.

Congenital heart disease occupies a leading place among a wide range of diseases that require the involvement of a pediatric cardiologist. The generally accepted minimum calculated indicator of the frequency of congenital heart disease is 8 cases per 1,000 live births.

Congenital heart disease is the most common congenital anomaly, occurring in almost 1% of live births. Among congenital defects, congenital heart disease is the leading cause of infant mortality.

Often in his practice, a doctor encounters the presence of several nosological forms in a patient's diagnosis. The combination of pathologies of different organs and systems in one diagnosis is quite understandable, since many diseases have a common etiopathogenetic basis.

Pediatric cardiology has always been the basis of pediatrics, simultaneously constituting its integration and having a significant impact on reducing perinatal mortality; which is the basis for developing an assessment of medicine in the country.

Congenital heart defects (CHD) consist of a wide range of anomalies and malformations affecting the heart and great vessels that develop in utero, are present at birth, and present to health care in infancy, adolescence, or adulthood. Cardiovascular anomalies typically result from defective morphogenesis during embryologic development. Malformations may be limited to the cardiovascular system (nonsyndromic) or occur in combination with anomalies of other systems as part of specific syndromes (syndromic). The most common CHDs diagnosed in infancy are muscular and perimembranous ventricular septal defects followed by secundum atrial septal defects, with a combined prevalence of 48.4 cases per 10,000 live births.

Approximately 85% of children have a multifactorial etiology of CHD in children, which is usually the only malformation in the child and is the result of the interaction of many individually undefined genes and a number of other causes. The risk of recurrence of CHD in a family varies depending on the cause. The risk is negligible for de novo mutations, 2–5% for nonsyndromic multifactorial CHD, and 50% for cases where the cause is an autosomal dominant mutation. It is important to identify genetic factors, as most patients with CHD survive to adulthood and potentially start families.

Anomalies in the anatomical development of the heart and large vessels usually form during the 2nd to 8th week of intrauterine development as a result of impaired embryonic morphogenesis and can be caused by both hereditary (gene, chromosomal, genomic, zygotic mutations) and environmental factors affecting the developing embryo. The specific causes of congenital heart disease are unknown. Most often,

congenital heart disease is sporadic, not associated with a syndrome, and of unclear etiology. Genetic etiology has not been identified in approximately 72% of patients with congenital heart disease. They are often associated with chromosomal abnormalities detected by karyotyping in more than 1/3 of patients with congenital heart disease. Most often, it is trisomy of chromosomes 21, 18 and 13. In addition to Down syndrome, there are about 20 hereditary syndromes, in most cases accompanied by congenital heart disease. In total, syndromic pathology is detected in 6-36% of patients. However, some of these anomalies account for only about 5-6% of patients with congenital heart disease. The monogenic nature of congenital heart disease has been proven in 8% of cases; about 90% are inherited multifactorially, i.e. they are the result of a combination of genetic predisposition and the influence of environmental factors. The latter act as provoking factors, revealing hereditary predisposition when the "threshold" of their combined action is exceeded. The risk of recurrence of congenital heart disease in a family varies depending on the cause. The risk is insignificant for newly emerged mutations, 2-5% for non-syndromic multifactorial congenital heart disease, and 50% when the cause is an autosomal dominant mutation. Defects of the genetic code and embryogenesis disorders can also be acquired - exposure of the fetus and the mother's body to certain unfavorable factors (radiation, alcoholism, drug addiction), endocrine diseases (diabetes mellitus, rubella, systemic lupus erythematosus, thyrotoxicosis), viral and other infections suffered in the first trimester of pregnancy (rubella, influenza, hepatitis B), taking medications (lithium preparations, warfarin, thalidomide, antimetabolites, anticonvulsants, lithium, isotretinoin, anticonvulsants). Mixed viral and enterovirus infections suffered by the fetus in utero are of great importance in the development of heart and vascular pathology. In addition to etiological factors, there are risk factors for the birth of a child with congenital heart disease. These include: maternal age; toxicosis and threat of termination of the first trimester of pregnancy; stillbirths in the anamnesis; the presence of children with congenital malformations in close relatives. It is unclear whether maternal age is an independent risk factor for the development of congenital heart disease. Paternal age may also be a risk factor.

The impact of unfavorable factors on a woman can disrupt the differentiation of the heart and lead to the formation of congenital heart disease. Such factors are considered to be:

• infectious agents (cytomegalovirus, herpes simplex virus, influenza virus, enterovirus, Coxsackie B virus, etc.);

• hereditary factors - in 57% of cases, congenital heart disease is caused by genetic disorders, which can occur both in isolation and as part of multiple congenital malformations; the most well-known causes of congenital heart disease are point gene

changes or chromosomal mutations in the form of deletion or duplication of DNA segments;

• somatic diseases of the mother, and primarily diabetes mellitus - lead to the development of hypertrophic cardiomyopathy and congenital heart disease;

• professional hazards and bad habits of the mother (chronic alcoholism, computer radiation, mercury and lead intoxication, exposure to ionizing radiation, etc.).

The most common defects are: ventricular septal defect - VSD (28.3%); atrial septal defect - ASD (10.3%); pulmonary artery stenosis (9.8%); tetralogy of Fallot - TF (9.7%); aortic stenosis (7.1%); coarctation of the aorta - PA (5.1%); transposition of the great vessels (4.9%); hypoplastic tricuspid valve syndrome, patent ductus arteriosus (PDA), and complete anomalous venous return are also encountered.

Intrauterine infectious pathology of the fetus and newborn is one of the most pressing and complex problems in pediatrics. The proportion of infectious and inflammatory processes in the structure of perinatal mortality is about 10-18%, second only to intrauterine asphyxia, respiratory disorders, and congenital anomalies. The relevance of the problem of intrauterine infection is due not only to significant peri- and postnatal losses, but also to the fact that children who have suffered a severe form of congenital infection very often develop serious health problems, leading to disability and a decrease in the quality of life in general.

Among the pathogens, a special position is occupied by TORCH complex infections - Toxoplasma (toxoplasmosis), Rubella (rubella), Cytomegalovirus (cytomegalovirus), Herpes (herpes). Includes such infections as hepatitis B and C, syphilis, chlamydia, gonococcal infection, HIV infection, listeriosis, enterovirus infection and others affecting the fetus. In the absence of adequate therapy during pregnancy, up to 50% of newborns with at least one TORCH infection in the mother are born with various injuries: intrauterine infections (IUI), intrauterine growth retardation (IUGR), hypotrophy, expected malformation (AM), including congenital heart disease, cerebrovascular accidents. With mixed infection, the incidence of newborns reaches 50-100%. In the studies of Lobzova A.V. (2014), immunological markers for a particular infection or association of infectious agents were diagnosed in newborns with and without congenital malformations. In this case, class G antibodies to viral-viral associations (CMV + herpes simplex virus (HSV)) were frequently determined both in the main and control groups (56.33 and 65.38%, respectively). In the main group, class G immunoglobulins to HSV were detected in 98.6% of the examined newborns and in 80.7% of the children in the control group, to CMV - in 95.8 and 96.1%, respectively, which is consistent with the literature data on the ever-increasing incidence of these pathogens in the population. Due to the fact that the frequency of intrauterine infections does not tend to decrease and, accordingly, there

are no adverse consequences, further study in this area is required to develop measures to prevent them.

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