AJPMHS GLOBAL RESEARCH NETWORK

AMERICAN Journal of Pediatric Medicine and Health Sciences
Volume 2, Issue 5, 2024 ISSN (E): 2993-2149

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 United States, which ranges from 4 to 50 per 1000 births, with the frequency of moderate and severe forms of congenital heart disease averaging 6%.

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

To date, more than 90 anatomical variants of congenital heart disease and about 200 different combinations of them have been described in domestic and foreign literature. In different regions of the country, heart and large vessel defects occupy 1-2 places in the structure of all congenital malformations, 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 healthcare system in the country or the diagnostic methods available to the doctor.

Congenital heart pathology occupies a major place among a wide range of diseases that require the involvement of a pediatric cardiologist. The generally accepted minimum estimated rate of congenital heart disease is 8 cases per 1 thousand live births.

CHD is the most common congenital anomaly, occurring in almost 1% of live births [11]. Among birth defects, congenital heart disease is the leading cause of infant mortality.

Often in his practice, a doctor is faced with 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 the reduction of perinatal mortality; which is the basis for the development of medical assessment in the country.

CHD consists 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 medical attention during infancy, adolescence, or adulthood. Cardiovascular abnormalities usually result from defective morphogenesis during embryological development. Malformations may be limited to the cardiovascular system (non-syndromic) or occur in combination with anomalies of other systems as part of certain syndromes (syndromic).

The most common congenital heart defects diagnosed in infancy are muscular and perimembranous ventricular septal defects followed by secondary atrial septal defects, with an overall prevalence of 48.4 cases per 10,000 live births.

About 85% of children have a multifactorial etiology of congenital heart disease in children, which is usually the only developmental defect in a child and is the result of the interaction of many, individually not yet identified genes and a number of other reasons. The risk of recurrence of congenital heart disease in a family varies depending on the cause. The risk is negligible for new onset mutations, 2–5% for non-syndromic multifactorial congenital heart disease, and 50% for cases where the cause is an autosomal dominant mutation. It is important to determine genetic factors, since most patients with congenital heart disease live to adulthood and potentially start families.

Anomalies of the anatomical development of the heart and large vessels usually form in the 2-8th week of intrauterine development as a result of disturbances in 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 not known. Most often, congenital heart disease is sporadic, not associated with a syndrome and of unclear etiology. A genetic etiology is not 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 this is trisomy on chromosomes 21, 18 and 13. In addition to Down's disease, there are about 20 hereditary syndromes, in most cases accompanied by congenital heart disease. In total, syndromic pathology is found 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. are the result of a combination of genetic predisposition and environmental factors. The latter act as provoking agents, revealing a hereditary predisposition when the "threshold" of their joint action is exceeded. The risk of recurrence of congenital heart disease in a family varies depending on the cause. The risk is negligible for new mutations, 2-5% for non-syndromic multifactorial congenital heart disease, and 50% when the cause is an autosomal dominant mutation. [eleven]. Defects of the genetic code and disorders of embryogenesis can also be acquired - the impact on the fetus and the mother's body of certain unfavorable factors (radiation, alcoholism, drug addiction), endocrine diseases (diabetes mellitus, rubella, systemic lupus erythematosus, thyrotoxicosis), viral and other infections transferred to I trimester of

pregnancy (rubella, influenza, hepatitis B), taking medications (lithium drugs, warfarin, thalidamide, antimetabolites, anticonvulsants, lithium, isotretinoin, anticonvulsants). Mixed viral and enteroviral infections transmitted by the fetus in utero are of great importance in the occurrence of pathology of the heart and blood vessels. In addition to etiological ones, risk factors for having a child with congenital heart disease are identified. These include: maternal age; toxicosis and threat of termination of the first trimester of pregnancy; history of stillbirths; the presence of children with congenital malformations in close relatives. It is unclear whether maternal age is an independent risk factor for developing congenital heart disease. Paternal age may also be a risk factor.

Exposure of a woman to unfavorable factors can impair cardiac differentiation and lead to the formation of congenital heart disease. These factors are considered to be:

- infectious agents (cytomegalovirus, herpes simplex virus, influenza virus, enterovirus, Coxsackie B virus, etc.);
- hereditary factors - in 57% of cases of congenital heart disease are caused by genetic disorders, which can occur either alone or as part of multiple congenital malformations; the most well-known causes of CHD 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;
- occupational hazards and bad habits of the mother (chronic alcoholism, computer radiation, mercury, lead intoxication, exposure to ionizing radiation, etc.).

The most common defects are the following: ventricular septal defect - VSD (28.3%); atrial septal defect - ASD (10.3%); pulmonary stenosis (9.8%); tetralogy of Fallot – TF (9.7%); aortic stenosis (7.1%); coarctation of the aorta - SPA (5.1%); transposition of the great vessels (4.9%); Tricuspid valve hypoplasia 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 share 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 quality of life in general.

Among pathogens, a special position is occupied by infections of the TORCH complex - Toxoplasma (toxoplasmosis), Rubella (rubella), Cytomegalovirus (cytomegalovirus), Herpes (herpes). Includes infections such as hepatitis B and C, syphilis, chlamydia, gonococcal infection, HIV infection, listeriosis, enterovirus infection and others that affect 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 damages: intrauterine infections (IUI), intrauterine growth retardation (IUGR), malnutrition, birth defects), including congenital heart disease, cerebrovascular accidents. With mixed infection, the morbidity rate in 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 newborn children with congenital malformations and without congenital defects. At the same time, class G antibodies to viral-viral associations (CMV + herpes simplex virus (HSV)) were often detected in both the main and control groups (56.33 and 65.38%, respectively). Moreover, in the main group, class G immunoglobulins to HSV were detected in 98.6% of examined newborns and in 80.7%) of

children in the control group, to CMV - in 95.8 and 96.1%, respectively, that is consistent with literature data on the ever-increasing damage to the population by these pathogens.

Due to the fact that the frequency of intrauterine infections does not tend to decrease and, accordingly, adverse consequences, further study in this direction is required to develop measures to prevent them.

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