AJPMHS GLOBAL RESEARCH NETWORK

AMERICAN Journal of Pediatric Medicine and Health Sciences
Volume 01, Issue 04, 2023 ISSN (E): XXX-XXX

Epidemiological and Etiological Data of Morphogenesis and Pathomorphology of Congenital Heart Diseases in Children

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Abstract: The article is devoted to the study of epidemiological and etiological data of morphogenesis and pathomorphology of congenital heart defects in children.
Modern data on the etiology and pathophysiological mechanisms of the development of this pathology are considered. A brief review of the literature on this issue was carried out.
Key words: epidemiology, etiology, congenital heart defects, children.

Relevance. Congenital malformation is currently considered as an anomaly of formation, which entailed gross changes in the structure and function of an organ or tissue that arose in the prenatal period. Among the various types of congenital and hereditary pathology: teratogenic defects account for up to 3%, intrauterine diseases - 2.5%, twinning - 0.5%, multifactorial diseases - 23%, genetic - 28% and diseases of unknown etiology - up to 43%. Approximately 2-3% of newborns have serious multiple congenital malformations Shermatova Z. A. et al. (2023)

Among non-inflammatory heart diseases in children, a certain place is occupied by congenital heart disease (CHD), which is formed as a result of a violation of embryogenesis at the 2-8th week of intrauterine development. Currently, an increase of up to 22% in the proportion of CHD, which play an important role in the formation of infant mortality and disability. According to most authors, the incidence of CHD ranges from 8 to 10 per 1000 births, with a tendency to increase in recent decades. The prevalence of congenital heart disease varies not only by years of observation, but also by region and with the age of children. In the first months of life, especially in the neonatal period, not all defects are diagnosed in a timely manner, at the same time they often lead to death, while others are detected later, determining the dynamics of prevalence and structure in different age periods. It is known that there are more than 90 variants of the IPU and many of their combinations. Without radical correction, 50-60% of children die in the first year of life. Mortality from heart defects is especially high in the neonatal period. According to statistics, every year in Kazakhstan, several thousand young children born with this pathology, 36% of them, if an urgent operation was not performed, die for the first time in 30 days, another 35% within 6 months, most of the survivors subsequently become disabled (Lim M. et al. (2021)

Foreign researchers noted that the prevalence of extracardiac pathology in children with CHD is 12-30%, in adults with CHD - 45-60% [Dilber D., Malcić I. 2010]. The 12 generally accepted methods for measuring comorbidity that exist in the world, including in diseases of the cardiovascular system, are applicable to adult patients (CIRS system (Cumulative Illness Rating Scale, ICED scale, Charlson index, etc.), but are not applicable to children. In modern pediatrics, there are a number of concomitant nosologies in cardiosurgical pediatric patients that require additional diagnostic and therapeutic measures and the development of management tactics at various stages of medical care, such as metabolic pathology (mucopolysaccharidosis, cystic

fibrosis, etc.), extracardiac malformations, and others. A report from the American Heart Association (AHA) notes that at least 40,000 children with congenital heart disease are expected in the United States in 2019, which is 1% of all newborns. Of these, about 25% of live births, or 0.25%, will require invasive treatment during the first year of life. The increase in the proportion of children with CHD is also associated with modern methods of surgical treatment, which ensure the survival of children with almost all defects and, as a result, the rapid growth of the population of adolescents and adults with operated CHD.

Pnjoyan A.A. et al. (2021) Every year, up to 16,000 surgeries are performed in the Russian Federation for congenital heart defects (CHDs) in children. Surgical treatment of congenital heart disease in children is the most important direction in cardiac surgery, both in terms of the complexity of the surgical intervention and the management of patients in the postoperative period. The most common complications of cardiopulmonary bypass surgery are atelectasis, distelectasis, bronchitis, tracheobronchitis, and pneumonia. The incidence of these complications is up to 14%. In many ways, the development of these processes is a consequence of a systemic inflammatory response in the postoperative period. Medical rehabilitation of this cohort of patients includes the following activities: the use of apparatus physiotherapy, therapeutic massage, exercise therapy. The studies of a number of authors demonstrate the high efficiency of rehabilitation treatment in the prevention of complications from the respiratory system. Efficiency in creating a mucolytic effect was shown by the use of the postural drainage procedure. Due to the combination of vibration and percussion of the chest in young children, secretion mobilization in the airways is provoked. Further chest compression leads to cough initiation, which in turn facilitates sputum evacuation. We studied the results of treatment of 80 children operated on for congenital heart disease. The average age of the children (boys 52.5%, girls 47.5%) was 7.2 months, and the average body weight was 5.7 kg. The inclusion of the postural drainage technique in combination with percussion and vibration in the early postoperative period in the rehabilitation treatment complex leads to a decrease in the length of stay on artificial ventilation, the length of stay in the intensive care unit and in the hospital as a whole. Suard C, Flori A, Paoli F, Loundou A (2020) French authors consider the possibility of prenatal screening for congenital heart disease in the population. Congenital heart defects (CHD) are the most common congenital malformations, accounting for one third of all cases. In Europe, these malformations represent about thirty-six thousand live births per year, or a prevalence of about seven out of one thousand live births. IHD is the leading cause of death due to congenital malformations in the first year of life. In European countries, screening rates vary depending on the setting of screening programmes. The screening rate ranges from 17.9% in the absence of organized screening to 55.6% with systematic 2 or 3 ultrasounds. In France, three ultrasounds are recommended during pregnancy. According to some studies, the morbidity-mortality benefits of prenatal CHD screening appear to be inconsistent in terms of survival. However, among neonates with CAD, prenatal diagnosis appears to be associated with lower rates of preoperative risk factors for cardiac surgery. In addition, prenatal diagnosis may improve the prognosis of children in terms of morbidity, especially in terms of the level of neurocognitive development. Some CAD is associated with genetic abnormalities, and prenatal screening for these allows for genetic testing. The overall rate of prenatal detection of coronary artery disease is 71.5% in the southern region of France. The prenatal detection rate was 47.3% in the Paris area, and the EUROCAT study in 2009 reported detection rates ranging from 1% (Malta) to 42.5% (France). In Europe, such differences between countries can be explained by unequal access to health care and differences in the policy of organizing prenatal screening. In France, this difference can be explained by inconsistencies in design between studies. In addition,

the improvement in detection rates can be explained by the evolving guidelines of the CNEOF (National Conference on Obstetrics and Ultrasound of the Fetus) and the French College of Fetal Sonography (CFEF) requiring three images of the fetal heart during ultrasound performed in the second and third trimesters: four cavities and right and left ejection channels.

Diab NS, Barish S, Dong W (2021) Congenital heart disease (CHD) is the most common congenital malformation and the leading cause of death in it. Genetic etiology accounts for about 90% of CHD cases, but so far the molecular diagnosis remains unresolved in 55% of patients. Copy number variation and aneuploidy account for ~23% of cases overall, and high-throughput genomic technologies have identified additional types of genetic variation in CAD. The first CAD risk genotypes identified by high-throughput sequencing were de novo mutations, many of which occur in chromatin-modifying genes. Mouse models of cardiogenesis further support the damaging nature of chromatin mutations that modify CAD. Transferable mutations have also been identified by population-scale sequencing of CAD cohorts, and many transferable mutations are enriched in ciliary and Notch or VEGF pathway genes. Although the authors have come a long way in identifying the causes of CAD, more work is needed to end the diagnostic odyssey for all families of CAD. Complex genetic explanations for CAD are emerging, but increasingly sophisticated analysis strategies applied to very large CAD cohorts will be needed before they can be realized in providing molecular diagnoses to genetically unresolved patients.

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