

ANALYSIS OF THE SIGNIFICANCE OF MEDICAL AND SOCIAL FACTORS IN THE FORMATION OF CONGENITAL HEART DEFECTS IN CHILDREN

Guzaliya Khasanova

PhD, assistant of the Department of Family Medicine No. 1 of the Tashkent Pediatric Medical Institute

<https://doi.org/10.5281/zenodo.10807912>

Abstract. *Congenital heart defects are one of the most common congenital anomalies in children (30% of all congenital malformations); they are the third most common after congenital pathology of the musculoskeletal system and central nervous system. In this regard, it was of interest to study the most likely risk factors for the formation of congenital heart defects in children.*

Purpose of the study. To study the risk factors for the formation of congenital heart defects in children based on an analytical review of medical records - 56 children with congenital heart defects hospitalized in the cardiology department of the National Children's Medical Center of the Republic of Uzbekistan.

Materials and methods. Analytical review of medical documentation (56 patients with congenital heart defects hospitalized in the cardiology department of the National Children's Medical Center of the Republic of Uzbekistan. The control group consisted of 32 children without pathology of the cardiovascular system.

Results. Leading risk factors were identified: anemia (48.5%), chronic placental insufficiency (38%), threatened miscarriage (32.7%), toxicosis of the first half of pregnancy (22.3%) and acute viral infection in the early stages of pregnancy (21.3%).

Conclusion. The risk of developing congenital heart defects in many cases is due to a combination of hereditary predisposition with a multilateral pathological influence of internal and external environmental factors, the simultaneity of their impact, as well as the duration of exposure to the damaging factor, the number of damaging factors and the simultaneity of their effects.

Keywords: *children, congenital heart defects, risk factors*

Introduction. The first definition of congenital heart defects (CHD) was given back in 1971. According to S. C. Mitchell, "CHD is a structural anomaly of the heart or intrathoracic great vessels, which actually or potentially leads to functional impairment" [1]. This concept has a number of disadvantages. Firstly, it does not take into account the time of occurrence of the defect. Secondly, a number of anomalies of the intrathoracic vessels are not related to congenital heart disease (for example, persistent superior vena cava), although they are important during surgical intervention [2]. Thirdly, congenital heart disease does not include diseases such as cardiomyopathies and anomalies of the cardiac conduction system (Wolf-Parkinson-White phenomenon, long QT syndrome), which are "structural abnormalities" and "leading to functional disorders" [2]. In addition, the term congenital heart disease includes congenital defects not only of the intrathoracic sections of the great vessels (for example, coarctation of the abdominal aorta). Currently, the concept of congenital heart disease is given a broader meaning, namely congenital heart disease can be defined as an anatomical deformation of the heart or large vessels that

develops in utero, regardless of when it was discovered [3]. Currently, CHDs occupy a leading position in terms of prevalence compared to other developmental defects in children and remain the leading cause of death [4–6]. Moreover, a further increase in the prevalence of congenital heart disease is predicted. This may be partly due to improvements in diagnostic techniques associated with increased qualifications of ultrasound specialists and improved imaging techniques. For example, a report by the American Heart Association (AHA) notes that at least 40,000 children with congenital heart disease are expected to be born in the United States in 2017, which is 1% of newborns. Of these, about 25% of live births, or 2.4/1000, will require invasive treatments during the first year of life. The increase in the proportion of children with congenital heart disease is also facilitated by modern methods of surgical treatment, which ensure the survival of children with almost all defects [7] and, as a consequence, the rapid growth of the population of adolescents and adults with operated on congenital heart disease [6, 7, 9]. According to various data, the prevalence rates of congenital heart disease in children vary significantly and range from 4 to 50 cases per 1000 live births [9, 10]. This variability is explained by differences in the criteria for their assessment. Thus, the prevalence of pathology increases when taking into account children with small congenital heart defects (for example, with a bicuspid aortic valve, small patent ductus arteriosus, PDA). The incidence of moderate and severe forms of congenital heart disease among US children is about 6 cases per 1000 live births, increases to 19/1000 when children with a bicuspid aortic valve are included and to 75/1000 when point muscular ventricular septal defects (VSD) are included [9]. Over the last century, the world has seen an increase in the prevalence of congenital heart disease from 0.6 in 1930–1934. up to 9.1 cases per 1000 live births after 1995 [11]. Over the past 15 years, there has been some stabilization of the values of this indicator, and about 1.5 million children with congenital heart disease are born annually in the world [12]. According to the European Registry of Congenital Abnormalities and Twins (EUROCAT), in the period 2010–2014. the prevalence of all congenital heart diseases was 8.1, including severe manifestations - 2.2 per 1000 newborns [8]. The prevalence of severe congenital heart disease has also increased over time, which is likely due to improved diagnostic methods for congenital heart disease and the prevention of antenatal and infant mortality. Thus, A. J. Marelli et al. in 2007 published the results of a large study covering the period 1985–2000, which demonstrated an increase in the proportion of severe congenital heart disease in the pediatric population by 22% [13]. Obviously, a similar trend is observed at the present time [14]. Significant geographic variations in the prevalence of congenital heart disease among live births around the world have been found, with the highest observed in Asian countries at 9.3/1000 [11].

In China, the prevalence of congenital heart disease, as of 2009, was 8.2/1000 people, of which 6.7 were among live births, and up to 168.8 among stillbirths [15]. The most common CHDs were atrial septal defect (ASD) (34%), PDA (24%), and VSD (11%) [15]. In India in 2014, the incidence of congenital heart disease was 19/1000 newborns, including VSD (33%), ASD (19%), tetralogy of Fallot (16%) [13]. In Saudi Arabia (based on 1993–2003 data), the prevalence of congenital heart disease ranged from 2.1 to 10.7/1000 people, with VSD (30–40%), ASD (9–18%) and valve stenosis being the most common conditions. pulmonary artery (6–12%). Overall, the incidence of severe congenital heart disease was approximately 5.4/1000 live births per year [9]. In Europe, the prevalence of congenital heart disease in 2011 was 6.9/1000 live births, and in North America it was 8.2/1000 live births [11]. The most common congenital heart defects in children were VSD, ASD, pulmonary valve defects and PDA [7]. According to Russian authors (from

2008), congenital heart defects are found in 7–17 per 1000 newborns [18]. There are a large number of registers to record cases of congenital anomalies, including congenital heart disease. An example is the British and Irish Network of Congenital Anomaly Researchers (BINOCAR) [5,6], Canadian Congenital Anomalies Surveillance Network (CCASN) [20], Australian Congenital Anomalies Surveillance Network (CCASN) [20]. Congenital Anomalies Monitoring System, ACAMS) [10]. Currently, there are two international registry monitoring systems: The International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR or Clearinghouse) and EUROCAT [22]. ICBDSR includes 47 regional registries from 36 European countries, Asia, America and Africa [12] EUROCAT unites 35 regional registries from 21 European countries [13]. The causes of congenital heart defects are not completely clear. Potentially dangerous risk factors for the formation of the pathology of the fetal cardiovascular system are numerous and can be combined. These include congenital and acquired diseases of the mother and fetus, as well as teratogenic environmental factors. Of great importance in the occurrence of defects is hereditary pathology (chromosomal and gene mutations), infectious, primarily viral diseases [3]. Identification of risk factors for the birth of a child with congenital heart defects is important not only from a theoretical but also from a practical point of view in order to predict the risk of development and plan adequate preventive measures to reduce them. CHD is classified as a multifactorial disease, since in their etiology, along with genetic factors, a certain role is played by the presence of bad habits in the child's parents, maintaining an unhealthy lifestyle [8]. To date, there has been a redistribution in the ranking of risk factors for congenital heart defects. So, according to Mutafyan O.A. (2005), the leading risk factors for the birth of a child with CHD are: the age of the mother, endocrine disorders in spouses, toxicosis in the 1st trimester and threats of termination of pregnancy, a history of stillbirth, the presence of other children with congenital malformations, a woman taking endocrine drugs to preserve pregnancy. Safiullina A.R. refers to the leading risk factors for the birth of children with congenital heart defects: medical abortion in the history of the mother; burdened with cytomegalovirus, herpetic infections, ureaplasmosis, viral hepatitis B, chlamydia, syphilis of the parents; serial number of pregnancy 4 or more; anemia and exacerbations of chronic diseases of the mother during pregnancy; mothers smoking; level of education of parents; incomplete family in a child [5]. According to other researchers, the significance of occupational hazards as a risk factor in women has decreased, and SARS during the 1st trimester of pregnancy, high blood pressure, an increase in the number of children born out of wedlock, and maternal smoking during pregnancy are significant [6]. Risk factors for the birth of a child with CHD also include: the age of the mother over 35, toxicosis in the 1st trimester, ionizing radiation, the presence of other children in the family with congenital malformations [7]. The unfavorable ecological situation in the region also plays an important role in the occurrence of defects. In connection with the foregoing, the purpose of our study was to study the risk factors for the development of congenital heart defects in children according to hospitalization at the National Children's Medical Center of the Republic of Uzbekistan.

The aim of the study to study the risk factors for the formation of congenital heart defects in children based on an analytical review of medical records - 56 children with congenital heart defects hospitalized in the cardiology department of the National Children's Medical Center of the Republic of Uzbekistan.

Materials and methods. Analytical review of medical documentation (56 patients with congenital heart defects hospitalized in the cardiology department of the National Children's Medical Center of the Republic of Uzbekistan. The control group consisted of 32 children without pathology of the cardiovascular system.

Results and discussion. Of the more than 90 types of malformations and about 200 different combinations of them, about half are malformations with enrichment of the pulmonary circulation. The structure of CHD in observed children: ventricular septal defect - 30.7%, atrial septal defect - 18.2%, patent ductus arteriosus - 11.5%, combined defects - 5.6%, complex defects - 3.2%, stenosis pulmonary artery - 3.2%, tetralogy of Fallot - 2.9%, coarctation of the aorta - 2%, malformations of the aortic valve - 1.8%, transposition of the great vessels - 0.4%. Our data on the structure of congenital heart defects correspond to the literature data. In terms of age, children were distributed as follows: up to 1 year old - 5 children (10%), 1-3 years old - 14 (28%), 4-6 years old - 14 children (28%), and 7-14 years old - 12 children (24%). It should be noted that in recent years there has been a significant increase in patients with congenital heart defects, to a greater extent this applies to young children. On the one hand, this may be due to an improvement in the quality of diagnosis of congenital heart defects, their early detection, but on the other hand, with the deterioration of the environmental situation in the region and the growing influence of various adverse factors on the formation of defects (occupational hazards, infections, low living standards, population).

It is known that 90% of heart defects are of a multifactorial nature, i.e., the combined effect of hereditary and environmental factors plays a role in their occurrence, 8% of them are caused by chromosomal abnormalities or a single gene defect, 2% are exclusively environmental factors (physical, chemical, biological). CHD are often detected among the syndromes of chromosomal diseases (in 3.2%): with trisomy 13 - Patau syndrome and trisomy 18 - Edwards syndrome, CHD are detected in 100%, with trisomy 21 - Down syndrome in 40-50% of cases, CHD is diagnosed, with Shereshevsky-Turner syndrome CHD occur in 20% of cases [2]. According to the cardiology department, one fifth of children with congenital heart defects have hereditary syndromes. Thus, in the CHD register there are 23 children with Down syndrome and 5 children with a Down-like phenotype, 2 children with Patau syndrome and 2 children with Edwards syndrome, one child each with Shereshevsky-Turner syndrome, Klinefelter syndrome, Christ-Stevens syndrome - Touraine. In 2.6% of cases, congenital malformations (septal malformations) were detected in the closest relatives of children with congenital heart defects. A detailed analysis of the parents' obstetric and somatic history made it possible to identify risk factors for the development of congenital heart defects in children in the Amur Region. We noted that half of the mothers had a complicated course of pregnancy. In the main group, pregnancy proceeded with the threat of termination in 32.7% of cases ($p < 0.05$), and in the control group in 11.7%. Toxicosis of the first half of pregnancy suffered in the main group 32.3% of pregnant women ($p < 0.05$), and in the control group only 13.3%. According to our data, chronic placental insufficiency (38%) is two times more in the main group than in the control group (18.5%). In half of the cases (48.5%) in mothers of the main group, pregnancy proceeded against the background of anemia ($p < 0.05$), and in the control group only in 12.7% of cases. Acute respiratory viral infection in the main group suffered 32.5% of pregnant women ($p < 0.05$), of which in the first half of pregnancy 21.3% and in the second half - 11.2% of mothers, and in the control group only in 13.4 %.

It should be noted that the earlier the fetus is affected, the more serious the pathological changes. The presence of one viral disease is still not enough for a future child to develop heart disease, however, subject to additional factors (the severity of the viral and bacterial disease, the presence of a genetic predisposition to adverse reactions to the triggering effect of this factor), the viral agent may be decisive in terms of the formation of congenital heart disease in a newborn [3]. A burdened obstetric history was observed in both groups. Previous miscarriages and abortions were noted in 21.5% of mothers of the main group and 19.4% in the control group, infertility was 4% in each group, and a history of stillbirth was 1.8% in both groups. When studying bad habits, it was revealed that in the main group 5.7% of children were born from fathers who abused alcohol during the conception of a child and 20.8% of pregnancies occurred against the background of passive nicotine intoxication, which is 4 times more often than in the control group ($p < 0.05$), since in the control group only 0.8% of fathers abused alcohol, and only 5.6% of mothers were exposed to passive smoking before pregnancy or during pregnancy. After analyzing the somatic pathology of mothers, it was revealed that in the main group, pregnancy more often occurred against the background of diseases of the genitourinary sphere (27.6%) (cystitis, colpitis, cervical erosion, chronic pyelonephritis), pathology of the cardiovascular system was less common (neurocirculatory dystonia due to hypotonic or hypertensive type, hypertension, coronary heart disease) (in 17.3%), endocrine system (euthyroidism, goiter, diabetes mellitus, obesity) (14.3%), digestive system (chronic gastritis and gastroduodenitis, duodenal ulcer intestines) (7.1%), respiratory organs (bronchitis, chronic and acute pneumonia) (5.1%), ENT organs (chronic sinusitis, tonsillitis, chronic tonsillitis) (4.1%). In the control group, somatic pathology was detected less frequently than in the main group and was represented by pathology of the genitourinary sphere (9.8%), endocrine system (6.8%), cardiovascular system (1.9%) and ENT pathology. (0.9%). Fathers had chronic diseases (chronic pyelonephritis, gastric ulcer, diabetes mellitus, bronchial asthma, upper respiratory tract diseases) in 9.6% in the main and 6.7% in the control group, and the rest considered themselves healthy. Most often in our country, children have septal defects (defects of the interventricular and interatrial septum, open ductus arteriosus). Among the probable risk factors for the development of congenital heart defects in children, the leading ones are: anemia in the mother during pregnancy (48.5%), chronic placental insufficiency (38%), the threat of abortion (32.7%), toxicosis of the first half of pregnancy (22.3%), acute viral infection in early pregnancy (21.3%), nicotine intoxication (20.8%). Among the somatic pathology, diseases of the genitourinary system (27.6%), the cardiovascular system (17.3%), and the endocrine system (14.3%) were noted more often.

Conclusion. The risk of developing congenital heart defects in many cases is due to a combination of hereditary predisposition with a multilateral pathological influence of internal and external environmental factors, the simultaneity of their impact, as well as the duration of exposure to the damaging factor, the number of damaging factors and the simultaneity of their effects. The main areas of prevention of the birth of children with congenital heart defects are the elimination of manageable risk factors for the formation of this defect, which includes the identification and timely treatment of anemia, chronic placental insufficiency, the threat of abortion, pregnancy toxicosis, the prevention and treatment of acute viral infections, the rehabilitation of chronic diseases of the pregnant woman and the refusal from smoking. Pregnant women with a risk of congenital heart disease in the fetus need close attention and screening, fetal echocardiography, consultation and observation of a pediatric cardiologist from the intranatal period. Carrying out

preventive measures aimed at eliminating these factors can significantly reduce the risk of having a child with congenital heart disease.

REFERENCES

1. Кардиология детского возраста / Под ред. Царегородцева А.Д., Белозерова Ю.М., Брегель Л.В. — М.: ГЭОТАР-Медиа; 2018 — 784 с. [Kardiologiya detskogo vozrasta. Ed by Tsaregorodtsev A.D., Belozerov Yu.M., Bregel' L.V. Moscow: GEOTAR-Media; 2014. 784 p. (In Russ).]
2. Клинические рекомендации по ведению детей с врожденными пороками сердца / Под ред. Бокерия Л.А. — М.: НЦССХ им. А.Н. Бакулева; 2021. — 342 с. [Klinicheskie rekomendatsii po vedeniyu detei s vrozhdennymi porokami serdtsa. Ed by Bokeriya L.A. Moscow: NCCSSH im. A.N. Bakuleva; 2021. 342 p. (In Russ).]
3. Alenezi AM, Albawardi NM, Ali A, et al. The epidemiology of congenital heart diseases in Saudi Arabia: a systematic review. *J Pub Health Epidemiol.* 2019;7(7):232–240. doi: 10.5897/JPHE2015.0723.
4. Bhardwaj R, Rai SK, Yadav AK, et al. Epidemiology of congenital heart disease in India. *Congenit Heart Dis.* 2015;10(5):437–446. doi: 10.1111/chd.12220.
5. Cheung P. Y. et al. Outcomes of preterm infants with congenital heart defects after early surgery: defining risk factors at different time points during hospitalization // *Frontiers in Pediatrics.* 2021; 8:616659. <https://doi.org/10.3389/fped.2020.616659>.
6. Fisher JG, Bairdain S, Sparks EA, Khan FA, Archer JM, Kenny M, Edwards EM, Soll RF, Modi BP, Yeager S, Horbar JD, Jaksic T. Serious congenital heart disease and necrotizing enterocolitis in very low birth weight neonates. *J Am Coll Surg.* 2019;220(6):1018-1026.e1014. <https://doi.org/10.1016/j.jamcollsurg.2014.11.026>
7. Napuoja L. et al. Somatic growth in children with congenital heart disease at 10 years of age: Risk factors and longitudinal growth. *Early Human Development.* 2021;156:105349. <https://doi.org/10.1016/j.earlhumdev.2021.105349>
8. Haq, F. U., Jalil, F., Hashmi, S., Jumani, M. I., Imdad, A., Jabeen, M., ...&Atiq, M. (2021). Risk factors predisposing to congenital heart defects. *Annals of pediatric cardiology.* 2021;4(2), 117. <https://doi.org/10.1016/j.ijcard.2017.08.009>
9. Marelli AJ, Mackie AS, Ionescu-Ittu R et al. Congenital heart disease in the general population: changing prevalence and age distribution. *Circulation.* 2019;115(2):163–172. doi: 10.1161/ CIRCULATIONAHA.106.627224.
10. Safiullina A.R., Yakovleva, L.V. Analysis of risk factors of development of congenital septal heart diseases // the Modern problems of science and education. 2020;4 (Electronic magazine) URL: www.science-education.ru/104-6678.
11. Scahill CJ, Graham EM, Atz AM, Bradley SM, Kavarana MN, Zyblewski SC. Preoperative feeding neonates with cardiac disease. *World J Pediatr Congenit Heart Surg.* 2019;8(1):62–68. <https://doi.org/10.1177/2150135116668833>
12. Toms R, Jackson KW, Dabal RJ, Reebals CH, Alten JA. Preoperative trophic feeds in neonates with hypoplastic left heart syndrome. *Congenit Heart Dis.* 2018;10(1):36–42. <https://doi.org/10.1111/chd.12177>

13. Van der Linde D, Konings EE, Slager MA, et al. Birth prevalence of congenital heart disease worldwide: a systematic review and meta-analysis. *J Am Coll Cardiol.* 2022;58(21):2241–2247. doi: 10.1016/j.jacc.2022.08.025.