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MEDICAL SCIENCES

RESEARCH ON THE PREVALENCE OF CONGENITAL HEART DEFECT DIAGNOSIS

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Introduction Since the 1970s, the incidence of congenital heart defects diagnosed in children has nearly doubled (from 4.547 cases per 1,000 live births in 1970-1974 to 9.410 per 1,000 live births in 2010-2017)[1]. These data provide a wide range of opportunities for interpretation.

On one hand, improved access to healthcare systems leads to enhanced diagnostic capabilities, resulting in an increase in the detection of congenital heart defects. On the other hand, it is quite possible that these improved diagnostic capabilities do not fully explain the rise in cases of heart defects among newborns, and that the increase in diagnostic cases should be attributed to other factors. This second assertion is supported by the fact that the increase in diagnostic cases over time is not uniform across developed countries located in different parts of the world. For instance, in developed African countries, the incidence of congenital heart defects is the lowest (2.315 per 1,000 live births), while it is the highest in developed Asian countries (9.342 per 1,000 live births) [1]

Aim The heterogeneity of data obtained from different parts of the world indicates that this issue requires more detailed investigation. Following the implementation of global childhood immunization, congenital heart defects have become a leading cause of child mortality after birth injuries, asphyxia, and preterm birth. Congenital heart defects are relatively common, and their detection in newborns necessitates immediate surgical intervention as well as lifelong treatment

depending on the severity of the condition.

Establishing clear risk factors may lead to a reduction in the prevalence of congenital heart defects if these factors are modifiable. Alternatively, it could improve screening methods for early diagnosis during pregnancy, allowing expectant mothers the option to terminate the pregnancy if it becomes known that the child will have a defect, thus reducing the duration and quality of life for the child.

Materials and Methods To investigate the problem, a review of scientific articles from the PubMed and ScienceDirect platforms was conducted.

Results and Discussion According to a meta-analysis, the most commonly diagnosed congenital heart defects in newborns today are ventricular septal defect, atrial septal defect, and patent ductus arteriosus. In contrast, the prevalence of defects such as pulmonary artery stenosis and Tetralogy of Fallot is nine times lower. Compared to data from 1970-1974, the prevalence of severe defects such as Tetralogy of Fallot and pulmonary artery stenosis has remained unchanged, while the diagnosis of heart defects with a more favorable prognosis (ventricular and atrial septal defects) has increased nearly 2.5 times. However, since the 1990s, the number of diagnosed cases of heart defects has remained nearly constant until 2017 [2]. This meta-analysis examined data from countries with high and medium socio-demographic index values. No correlation was found between a country's economic well-being and the diagnosis of congenital heart defects; however, significant differences were noted in the frequency of detection of congenital heart defects based on the region of the world in which the country is located. For example, developed Asian countries emerged as leaders in the diagnosis of both severe and moderate congenital heart defects, while the lowest number of diagnosed defects was found in developed African countries.

Another meta-analysis presents interesting data, investigating the prevalence of diagnosed congenital heart defects and the mortality rate caused by these defects in relation to the country's socio-demographic index. It assessed data from 1990 to 2015. According to this analysis, the lowest detection rate of congenital heart defects was recorded in countries with a high socio-demographic index, while the highest

rates were found in countries with a low socio-demographic index [2]. The authors explain this phenomenon by the widespread availability of prenatal diagnostics, which allows for pregnancy termination upon the diagnosis of heart defects. Moreover, during this period, no increase in the prevalence of diagnosed congenital developmental defects was observed, aligning with the results of the previously mentioned meta-analysis.

Another notable observation is that the lower the socio-demographic index of a country, the greater the disparity in the frequency of congenital heart defect detection between boys and girls. In countries with a low socio-demographic index, the prevalence of congenital heart defects is 28 per 1,000 live births for boys and 21 per 1,000 live births for girls [2]. In developed countries, boys are more likely to be born with heart defects than girls, but the difference between these figures is significantly lower. It is evident that male gender is a risk factor for congenital heart defects, but this factor is nearly neutralized in developed countries due to the availability of prenatal diagnostics.

Data from both meta-analyses indicate that the frequency of diagnosed congenital developmental defects has remained nearly unchanged since the 1990s. However, returning to the data from the first meta-analysis, the question of the reasons behind the increase in diagnosed congenital heart defects between the 1970s and 1990s remains unresolved. To clarify this phenomenon, data presented by American researchers can be utilized. They compared cases of death caused by congenital heart defects in the 1980s and the early 2000s. In the first case, 49% of individuals who died due to congenital heart defects were under the age of 20 (27% during the first year of life). In the second case, this percentage had decreased to 9.3%. Most children who died in their first year of life were diagnosed with congenital heart defects only during autopsy. Currently, most patients with mild developmental defects who have undergone surgery have a positive life prognosis, and their life expectancy is nearly indistinguishable from the average in the population. For those with more severe heart defects, the prognosis varies depending on the severity of the condition and the adequacy of the medical care provided [3].

The authors explain the phenomenon of the increasing number of diagnosed heart defects in the second half of the 20th century by the rapid development of ultrasound diagnostics during this period, which significantly improved the detection of mild congenital heart defects such as atrial and ventricular septal defects, as well as patent ductus arteriosus. This aligns with the conclusions drawn by the authors of the first meta-analysis [1, 3]. At the same time, the frequency of diagnosing Tetralogy of Fallot and other severe heart defects has remained nearly unchanged in quantitative terms; however, the timing of their detection has shifted from the neonatal period to the prenatal period, allowing for the option of pregnancy termination upon their diagnosis. According to the authors, this, along with the improvement of diagnostic methods for milder heart defects and consequently the early initiation of treatment, has contributed to a nearly tenfold reduction in mortality among individuals with congenital developmental defects under the age of 20 [3].

Thus, the available data suggest that the increase in diagnosed cases of congenital heart defects can be attributed to advancements in diagnostic methods. The heterogeneity in the number of diagnosed heart defects among newborns, depending on the region of the world in countries with comparable socio-demographic indices, is explained by genetic factors, although these conclusions are somewhat questionable. Epidemiological studies assert that only 15% of cases can be explained by known genetic risk factors and up to 2% by known environmental factors (which include pollution levels in air and water, as well as maternal smoking and diseases contracted during pregnancy). The etiology remains unknown for more than 80% of diagnosed congenital developmental defects [4, 5].

Conclusions. The increase in the diagnosis of congenital heart defects in newborns can be attributed to improvements in instrumental diagnostic methods, primarily the widespread use of ultrasound examinations. At the same time, there has been a reduction in childhood mortality due to congenital heart defects, thanks to both antenatal diagnosis and prenatal screening. However, the fact that the level of diagnosis of heart defects among newborns has remained unchanged since the 1990s indicates that there has been no improvement in either the quality of prenatal

screening or the understanding of the etiological factors of congenital heart defects since that time. Currently, we can identify the cause of congenital heart defects in less than 20% of cases. There is a clear need to research the etiological factors of congenital heart defects; this would enable more effective prenatal diagnosis, prevention, and treatment, and reduce the burden of disease for both parents and the child, as well as for the state.

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