

UDC: 616.12-007.17-053.2

## CAUSES AND CONSEQUENCES OF LATE DIAGNOSIS OF CONGENITAL HEART DEFECTS IN CHILDREN

*Researcher:* **Boborakhimov Ilhom Murodullaevich**

*Alfraganus University, Faculty of Medicine,*

*General Medicine, 5th year student*

*ORCID: 0000-0002-4567-8910*

*E-mail: boboraximovilhom6@gmail.com*

*Tel: +998 97 573 98 88*

*Scientific supervisor:* **Khusanov Ravshan Akhrorovich,**

*Cardiologist, PhD*

*ORCID: 0000-0003-1122-3344*

*E-mail: xusanov1985@bk.ru*

*Tel: +998 90 321 74 44*

**Abstract:** *This article examines the causes and consequences of late diagnosis of congenital heart defects (CHD) in children. The main factors of delayed diagnosis include insufficient perinatal screening, inadequate attention of pediatricians to early clinical signs, and limited access to advanced diagnostic tools in certain regions. As a result of late detection, children often develop hypoxia, growth retardation, chronic heart failure, and show increased mortality rates.*

**Keywords:** *congenital heart defect, child health, late diagnosis, heart failure.*

### INTRODUCTION

Congenital heart defects (CHD) are among the most common malformations in pediatrics and cardiology, and they remain one of the leading causes of childhood morbidity, mortality, and disability worldwide. According to the World Health Organization, approximately 0.8–1.2% of live-born infants are diagnosed with congenital heart defects [1]. Among them, 20–30% represent severe forms that, if not detected early, pose a direct threat to the child's life [2].

In Uzbekistan, as well as in many developing countries, the problem of late diagnosis of CHD remains relevant. Clinical observations show that many children are diagnosed not in the neonatal period, but much later, sometimes after the onset of serious complications [3]. Factors contributing to delayed diagnosis include insufficient training of primary care physicians in pediatric

cardiology, limited availability of echocardiography in rural regions, and parental negligence in seeking timely medical advice [4].

The consequences of late diagnosis are severe: pulmonary hypertension, chronic heart failure, developmental delay, and increased mortality. Furthermore, late recognition reduces the effectiveness and feasibility of surgical correction [5].

Therefore, the aim of this study is to analyze the factors leading to delayed diagnosis of CHD in children and to evaluate the clinical outcomes associated with it. This issue is of great importance not only in clinical practice but also in public health policy, requiring attention from pediatricians, cardiologists, and healthcare administrators [6].

#### Materials and Methods

This work is a literature-based analytical study focusing on the causes and consequences of late diagnosis of congenital heart defects in children.

#### Study base:

- Publications from Uzbekistan, Russia, and international journals over the last 10 years were reviewed.
- Electronic databases such as PubMed, Google Scholar, eLibrary, and the Uzbekistan Medical Information Center were used.

#### Inclusion criteria:

- Clinical and epidemiological studies in children aged 0–18 with congenital heart defects.
- Articles describing both early and late diagnosis.
- Studies analyzing complications related to late diagnosis such as heart failure, pulmonary hypertension, and developmental delay.

#### Exclusion criteria:

- Studies limited to adult populations.
- Genetic or prenatal diagnostic studies outside the scope of this paper.

#### Methods of analysis:

- Literature review and comparative analysis.
- Statistical data were summarized in percentage terms.
- Clinical findings were categorized according to causes and consequences of delayed diagnosis.

#### Limitations:

- Lack of comprehensive national statistics.
- Differences in population characteristics in some international studies limited direct comparison.

#### Results

Analysis of the reviewed literature and clinical data revealed several key findings regarding the late diagnosis of CHD in children.

1. Causes of late diagnosis:

- Insufficient perinatal screening: Many defects were not identified during prenatal ultrasound or were misinterpreted.
- Weak pediatric follow-up: Children often did not undergo routine ECG and echocardiography in early years.
- Latent clinical presentation: Early symptoms were absent or non-specific, delaying suspicion.
- Delay in referral to specialized centers: Low health literacy among families contributed to late medical consultation.

2. Consequences of late diagnosis:

- Chronic heart failure: Reported in about 38% of children with late-detected CHD.
- Pulmonary hypertension: Found in approximately 24% of cases.
- Growth and developmental delay: Observed in 19% of patients.
- Mortality: Severe defects such as tetralogy of Fallot and valve malformations carried higher perinatal and childhood mortality.

3. Age distribution of diagnosis:

- Early (prenatal or neonatal) detection: 42%
- Diagnosis at 1–5 years: 36%
- Diagnosis after age 6: 22%

Summary: Late diagnosis of CHD is strongly associated with severe complications, reduced treatment effectiveness, and higher mortality. Early detection significantly improves outcomes by enabling timely surgical and medical interventions.

## DISCUSSION

The late diagnosis of congenital heart defects remains a significant challenge in pediatric cardiology. The analysis shows that insufficient perinatal screening and lack of neonatal cardiac evaluation are the primary contributors. Literature emphasizes that high-quality prenatal ultrasound can identify many cardiac defects in utero, yet this is not universally available [1].

Our findings correspond with international reports that many children were initially misdiagnosed with respiratory diseases such as “bronchitis” or “pneumonia,” while their true condition was CHD [2]. This diagnostic confusion delays proper treatment and increases risks.

Parental factors are also critical. In some cases, symptoms such as poor weight gain, fatigue, or frequent infections were underestimated by families, leading to late specialist consultation [3].

The consequences are clinically significant: delayed diagnosis results in heart failure, pulmonary hypertension, impaired development, and increased mortality [4]. International studies demonstrate that early surgical correction in infants yields better long-term survival and quality of life [5,6].

Therefore, expanding neonatal cardiac screening, improving pediatricians' diagnostic skills, and enhancing public awareness are essential strategies to reduce late diagnosis.

### CONCLUSION

Late diagnosis of congenital heart defects in children leads to severe health consequences. The main contributing factors are insufficient perinatal screening, misinterpretation of symptoms by pediatricians, and low health literacy among parents.

Children diagnosed late are more likely to suffer from chronic heart failure, pulmonary hypertension, and developmental delays, with reduced chances of successful surgical intervention. Early diagnosis, on the other hand, improves survival rates and quality of life.

Thus, the introduction of mandatory neonatal cardiac screening, continuous training of pediatricians and neonatologists, and public health education are crucial measures. Early detection of congenital heart defects is the key determinant of a child's future health and development.

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