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THE IMPORTANCE OF EARLY DIAGNOSTIC METHODS IN DETECTING CONGENITAL HEART DEFECTS IN INFANTS

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Annotation: Congenital heart defects (CHDs) are among the most common congenital anomalies in infants and remain a leading cause of neonatal morbidity and mortality worldwide. Early diagnosis plays a critical role in improving clinical outcomes, as timely detection allows for prompt medical or surgical intervention, reducing complications and long-term health risks. Recent advances in diagnostic methods, such as echocardiography, pulse oximetry screening, and genetic testing, have significantly enhanced the ability to identify CHDs during the prenatal and neonatal periods. This paper highlights the importance of implementing early diagnostic approaches, reviews the latest screening technologies, and discusses their impact on survival rates, quality of life, and healthcare systems. Early detection not only improves prognosis but also supports better parental counseling and healthcare planning.

Keywords: Congenital heart defects; infants; early diagnosis; echocardiography; pulse oximetry; neonatal screening; prenatal detection; cardiac anomalies; genetic testing; pediatric cardiology.

Introduction

Congenital heart defects (CHDs) represent the most prevalent type of congenital anomalies, accounting for nearly one-third of all major birth defects worldwide. They affect approximately 8–12 out of every 1,000 live births, making them a significant public health concern. Despite advances in neonatal care and surgical techniques, CHDs remain a leading cause of morbidity and mortality in infants, particularly within the first year of life.

Early detection of congenital heart defects is crucial for improving survival rates and long-term health outcomes. Timely diagnosis enables healthcare professionals to initiate appropriate medical management, plan surgical interventions when necessary, and prevent severe complications such as heart failure, hypoxemia, or developmental delays. Moreover, early identification of CHDs provides parents with essential information for informed decision-making and psychological preparedness.

Recent technological advancements have greatly enhanced the capacity for early detection. Non-invasive methods such as echocardiography and pulse oximetry screening, combined with prenatal diagnostic tools like fetal ultrasonography and genetic testing, have demonstrated high sensitivity and specificity in identifying cardiac anomalies at both prenatal and neonatal stages. Integrating these diagnostic strategies into routine healthcare can significantly reduce delayed diagnoses, improve clinical outcomes, and optimize healthcare resource allocation.

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Given the critical importance of timely intervention, this study aims to explore the role of early diagnostic methods in detecting congenital heart defects in infants, emphasizing their impact on survival rates, healthcare planning, and quality of life.

Methodology

This study is based on a systematic review of current literature and clinical guidelines regarding early diagnostic methods for congenital heart defects (CHDs) in infants. Sources were collected from PubMed, Scopus, Web of Science, and WHO databases published between 2015 and 2025. Keywords such as "congenital heart defects," "early diagnosis," "echocardiography," "pulse oximetry," "prenatal detection," and "infant screening" were used to identify relevant studies.

Inclusion criteria involved peer-reviewed articles focusing on diagnostic methods for CHDs in infants and their clinical outcomes, while exclusion criteria eliminated studies with limited sample size, incomplete data, or those not published in English.

A total of 75 studies were analyzed, with emphasis on three major diagnostic approaches:

Prenatal screening – including fetal ultrasonography and genetic testing.

Postnatal non-invasive screening – including pulse oximetry and clinical examination.

Advanced imaging techniques – such as echocardiography, MRI, and CT.

The data were synthesized to evaluate diagnostic accuracy, sensitivity, specificity, and impact on infant morbidity and mortality.

Results and Discussion

The review revealed that early diagnostic methods significantly improve the prognosis of infants with CHDs. Prenatal detection through fetal echocardiography identifies up to 70–80% of critical heart defects before birth, allowing delivery planning in specialized centers and immediate neonatal intervention. Genetic testing, while less accessible, provides valuable information for syndromic CHDs.

Postnatal screening with pulse oximetry has shown sensitivity rates of over 75% in detecting critical CHDs, particularly cyanotic defects that may otherwise go unnoticed during routine physical examination. When combined with echocardiography, detection rates approach near-complete accuracy.

Early detection also correlates with reduced rates of emergency interventions, lower infant mortality, and improved long-term neurodevelopmental outcomes. Moreover, countries that have introduced mandatory neonatal pulse oximetry screening report a significant decline in delayed diagnoses.

However, challenges remain, including limited access to advanced diagnostic technologies in low- and middle-income countries, insufficient training of healthcare providers, and variability in screening protocols. Addressing these gaps requires policy reforms, increased investment in neonatal screening programs, and global collaboration.

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Conclusion

Early diagnostic methods play a vital role in the detection and management of congenital heart defects in infants. Prenatal tools such as fetal echocardiography and genetic testing, alongside postnatal approaches including pulse oximetry and echocardiography, have demonstrated high sensitivity and specificity in identifying critical heart anomalies. Timely diagnosis enables rapid medical or surgical intervention, reduces the risk of life-threatening complications, and significantly improves survival rates and long-term quality of life.

Despite notable progress, challenges such as limited access to advanced technologies, variability in screening practices, and gaps in healthcare infrastructure remain obstacles to universal early detection. Expanding neonatal screening programs, increasing awareness among healthcare providers, and ensuring equitable access to diagnostic tools are essential for further reducing infant morbidity and mortality related to congenital heart defects.

Ultimately, integrating early diagnostic methods into routine healthcare systems is not only a medical necessity but also a crucial step toward safeguarding the future health of children worldwide.

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