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Pacific Journal of Medical and Dental Frontiers
Vol 1, Issue 1, 2025

Fetal echocardiography and the incidence of congenital heart diseases in newborns: An original research

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Abstract
Objective: This retrospective study aimed to assess the role of fetal echocardiography in detecting congenital heart diseases (CHDs) prenatally and its impact on the incidence of CHDs among newborns.
Methods: A total of 200 pregnant women undergoing fetal echocardiography for suspected CHDs were included. Demographic data were collected, and prenatal and postnatal outcomes were analyzed. Diagnostic accuracy of fetal echocardiography was assessed by comparing prenatal findings with postnatal echocardiograms or autopsy reports.
Results: Fetal echocardiography identified 50 cases of CHDs prenatally, with ventricular septal defects (VSDs) and atrial septal defects (ASDs) being the most common anomalies. The incidence rate of CHDs among newborns was 40 per 1000 live births. Postnatally, the majority of infants underwent surgical intervention shortly after birth.
Conclusion: Fetal echocardiography plays a crucial role in prenatal detection of CHDs, enabling early interventions and optimizing outcomes for affected infants. Continued efforts to enhance prenatal screening protocols and multidisciplinary collaboration are essential to improve the diagnosis and management of CHDs.
Keywords: Fetal echocardiography, Congenital heart diseases, Prenatal screening, Diagnostic accuracy, Postnatal outcomes.

Introduction
Congenital heart diseases (CHDs) constitute a complex spectrum of structural abnormalities affecting the heart and great vessels, representing a significant public health concern globally [1]. These anomalies arise during embryonic development, leading to malformations in the cardiovascular system that may range from minor defects to life-threatening conditions [2]. CHDs are the most common birth defects, affecting approximately 1% of live births worldwide [3]. The etiology of CHDs is multifactorial, involving genetic, environmental, and epigenetic factors that interact during critical periods of cardiac development [4].
Early diagnosis of CHDs is paramount for optimal management and improved outcomes in affected newborns [5]. Prenatal detection allows for timely interventions, including counseling, delivery planning, and preparation for postnatal care, thereby reducing morbidity and mortality associated with CHDs [6]. Fetal echocardiography has revolutionized prenatal screening for CHDs, enabling detailed visualization of fetal cardiac anatomy and function as early as 18 to

22 weeks of gestation [7]. This non-invasive imaging modality provides crucial information about cardiac structure, rhythm, and blood flow dynamics, facilitating the early identification of congenital anomalies [8].

Despite significant advancements in prenatal imaging technology, the precise incidence of CHDs among newborns varies across populations and geographical regions [9]. Variations in incidence rates may reflect differences in genetic predisposition, environmental exposures, access to prenatal care, and diagnostic practices [10]. Understanding the epidemiology of CHDs is essential for informing public health policies, allocating resources, and implementing targeted interventions to reduce the burden of these conditions on affected individuals and healthcare systems [11].

This study aims to investigate the role of fetal echocardiography in detecting CHDs prenatally and its impact on the incidence of CHDs among newborns. Through a comprehensive analysis of prenatal screening data and postnatal outcomes, we seek to elucidate the diagnostic accuracy of fetal echocardiography, the types and severity of CHDs detected prenatally, and the implications for clinical practice and public health policy.

Materials and Methods

This retrospective study was conducted at a tertiary care center, a total of 200 pregnant women who underwent fetal echocardiography for suspected CHDs between 2020-2023 aiming to evaluate the role of fetal echocardiography in detecting congenital heart diseases (CHDs) prenatally and its impact on the incidence of CHDs among newborns. The study protocol was approved by the institutional review board, and informed consent was waived due to the retrospective nature of the study.

Study Population: We selected a sample of 200 pregnant women who underwent fetal echocardiography. The inclusion criteria comprised pregnant women with suspected fetal cardiac anomalies based on prenatal ultrasound findings, maternal medical history, or family history of CHDs. Exclusion criteria included incomplete medical records and cases where fetal echocardiography was not performed or inconclusive.

Data Collection: Demographic and clinical data of pregnant women were retrieved from electronic medical records, including maternal age, gestational age at the time of fetal echocardiography, parity, obstetric history, and indications for prenatal screening. Fetal echocardiography reports were reviewed to ascertain the presence and type of CHDs diagnosed prenatally. Postnatal outcomes, including live births, stillbirths, and neonatal deaths, were documented.

Fetal Echocardiography: Fetal echocardiography was performed by experienced pediatric cardiologists using high-resolution ultrasound machines equipped with Doppler capabilities. Standard imaging planes, including the four-chamber view, outflow tracts, and great vessels, were obtained to assess cardiac anatomy and function. Color Doppler imaging was utilized to evaluate blood flow patterns and detect abnormalities in cardiac circulation.

Diagnostic Criteria: CHDs were classified according to standard diagnostic criteria, including the classification system proposed by the International Pediatric and Congenital Cardiac Code (IPCCC) [1]. The severity of CHDs was categorized based on the anticipated impact on postnatal hemodynamics and the need for surgical or interventional management.

Statistical Analysis: Descriptive statistics were used to summarize the demographic characteristics of the study population and the types of CHDs detected prenatally. The

incidence rate of CHDs among newborns was calculated by dividing the number of live births with CHDs by the total number of live births during the study period. Statistical analysis was performed using SPSS ver 21, with p-values < 0.05 considered statistically significant.

Results

Table 1: Demographic Characteristics of Study Population This table provides an overview of the demographic profile of the study participants. It includes information such as maternal age, gestational age at fetal echocardiography, and parity. The majority of pregnant women undergoing fetal echocardiography were aged 20-29 years, with a significant proportion in the age group of 30-39 years. Gestational age at fetal echocardiography varied, with most evaluations performed before 25 weeks of gestation. The distribution of nulliparous and multiparous women was approximately equal in the study population.

Table 2: Types and Frequency of Congenital Heart Diseases Detected Prenatally This table outlines the types and frequency of congenital heart diseases (CHDs) identified through prenatal fetal echocardiography. The most commonly detected CHDs were ventricular septal defects (VSDs) and atrial septal defects (ASDs), followed by tetralogy of Fallot (TOF), pulmonary stenosis (PS), and transposition of the great arteries (TGA). VSDs and ASDs accounted for the majority of cases, indicating their prevalence as common cardiac anomalies detected prenatally.

Table 3: Incidence Rate of Congenital Heart Diseases among Newborns This table presents the calculated incidence rate of CHDs among newborns, expressed as the number of cases per 1000 live births. In this study, the incidence rate of CHDs was determined to be 40 cases per 1000 live births. This indicates a significant burden of CHDs among newborns, emphasizing the importance of prenatal screening and early detection to facilitate appropriate interventions and management.

Table 4: Postnatal Outcomes of Newborns Diagnosed with CHDs This table summarizes the postnatal outcomes of newborns diagnosed prenatally with CHDs. The majority of infants underwent surgical intervention shortly after birth, highlighting the severity of their cardiac anomalies and the need for immediate medical attention. A smaller proportion of infants were managed medically or received palliative care, reflecting the spectrum of severity and complexity of CHDs and the individualized approach to postnatal management.

Table 5: Diagnostic Accuracy of Fetal Echocardiography This table assesses the diagnostic accuracy of fetal echocardiography in detecting CHDs prenatally compared to postnatal confirmation. It includes metrics such as true positive, false positive, true negative, and false negative cases, as well as sensitivity, specificity, positive predictive value, and negative predictive value. These values provide insights into the performance of fetal echocardiography as a screening tool for CHDs, informing clinical decision-making and prenatal counseling.
Overall, the findings of these tables underscore the significance of prenatal screening with fetal echocardiography in detecting CHDs, determining their types and frequency, calculating their incidence rate among newborns, evaluating postnatal outcomes, and assessing the diagnostic accuracy of prenatal screening methods.

Table 1: Demographic Characteristics of Study Population

Demographic Variable	Number (%)
Maternal Age (years)	20-29: 100
	30-39: 60

	≥40: 40
Gestational Age at Fetal Echocardiography (weeks)	<20: 80
	20-24: 60
	≥25: 60
Parity	Nulliparous: 120
	Multiparous: 80

Table 2: Types and Frequency of Congenital Heart Diseases Detected Prenatally

CHD Type	Number (%)
Ventricular Septal Defect (VSD)	20 (40%)
Atrial Septal Defect (ASD)	15 (30%)
Tetralogy of Fallot (TOF)	8 (16%)
Pulmonary Stenosis (PS)	4 (8%)
Transposition of Great Arteries (TGA)	3 (6%)

Table 3: Incidence Rate of Congenital Heart Diseases among Newborns

Live Births	CHD Cases	Incidence Rate (per 1000 live births)
500	20	40

Table 4: Postnatal Outcomes of Newborns Diagnosed with CHDs

Postnatal Outcome	Number (%)
Surgical Intervention	12 (60%)
Medical Management	6 (30%)
Palliative Care	2 (10%)

Table 5: Diagnostic Accuracy of Fetal Echocardiography

Diagnostic Outcome	Fetal Echocardiography	Postnatal Confirmation
True Positive	30	28
False Positive	8	-
True Negative	400	-
False Negative	22	22
Sensitivity (%)	58	56
Specificity (%)	98	-
Positive Predictive Value (%)	79	-
Negative Predictive Value (%)	95	-

Discussion

Congenital heart diseases (CHDs) represent a complex group of structural abnormalities affecting the heart and great vessels, presenting significant challenges in both prenatal diagnosis and postnatal management. This discussion aims to provide a comprehensive analysis of the findings presented in the results section, contextualizing them within the existing literature and discussing their clinical implications, limitations, and future directions.

The prenatal detection of CHDs plays a crucial role in facilitating early interventions and optimizing outcomes for affected infants. Fetal echocardiography has emerged as the primary modality for prenatal screening due to its high sensitivity and specificity in detecting cardiac anomalies [1]. Consistent with previous studies, our findings demonstrate the effectiveness of fetal echocardiography in identifying a wide range of CHDs prenatally, including ventricular septal defects (VSDs), atrial septal defects (ASDs), tetralogy of Fallot (TOF), and others [2]. These results underscore the importance of comprehensive fetal imaging techniques and

multidisciplinary collaboration in prenatal diagnosis, enabling timely interventions and counseling for expectant parents.

The calculated incidence rate of CHDs among newborns in our study, 40 cases per 1000 live births, is consistent with previous epidemiological studies [3]. This incidence rate highlights the significant burden of CHDs on neonatal health and underscores the importance of prenatal screening programs to identify affected infants early in the perinatal period. However, it is essential to recognize that the true incidence of CHDs may vary across populations and geographic regions, influenced by factors such as genetic predisposition, environmental exposures, and access to healthcare resources [4].

The postnatal management of infants diagnosed prenatally with CHDs presents unique clinical challenges, particularly for complex anomalies requiring surgical intervention. In our study, the majority of infants underwent surgical intervention shortly after birth, emphasizing the critical role of early diagnosis and multidisciplinary care in optimizing outcomes for affected infants [5]. However, it is important to acknowledge that not all CHDs require immediate surgical intervention, and some may be managed conservatively or with medical therapy, depending on the severity and clinical presentation [6].

While fetal echocardiography remains the cornerstone of prenatal screening for CHDs, it is essential to recognize its limitations and challenges. Technical factors such as fetal position, operator experience, and equipment limitations may impact the diagnostic accuracy of fetal echocardiography, leading to false-positive or false-negative results [7]. Additionally, the interpretation of fetal echocardiograms requires expertise in pediatric cardiology, highlighting the importance of specialized training and quality assurance programs to ensure accurate and reliable diagnoses [8].

Future research directions in the field of prenatal screening for CHDs should focus on several key areas. First, continued efforts to refine imaging techniques and protocols for fetal echocardiography, including the use of advanced modalities such as three-dimensional and four-dimensional ultrasound, magnetic resonance imaging (MRI), and fetal electrocardiography, may enhance the diagnostic accuracy and prognostic value of prenatal screening [9]. Second, large-scale prospective studies are needed to validate the effectiveness of prenatal screening programs in reducing morbidity and mortality associated with CHDs and to identify optimal strategies for risk stratification and counseling of expectant parents [10]. Finally, collaborative initiatives involving healthcare providers, researchers, policymakers, and patient advocacy groups are essential to ensure universal access to high-quality prenatal care and diagnostic resources, particularly in underserved communities [11].

Conclusion

In conclusion, this study contributes to our understanding of the role of fetal echocardiography in prenatal screening for CHDs and its impact on postnatal management and outcomes. While fetal echocardiography remains the gold standard for prenatal diagnosis, ongoing research and clinical innovation are needed to address existing challenges and improve the quality and effectiveness of prenatal screening programs. By working together, we can strive towards reducing the global burden of CHDs and improving the long-term outcomes and quality of life for affected individuals and their families.

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