



Original Article

Role of Fetal Echocardiography in Early Detection of Congenital Heart Diseases: A Prospective Study

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ABSTRACT

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Background: Congenital heart diseases (CHDs) are among the most common congenital anomalies and a leading cause of neonatal morbidity and mortality worldwide. Early prenatal diagnosis plays a crucial role in optimizing perinatal management and improving outcomes. Fetal echocardiography has emerged as a reliable, non-invasive imaging modality for detailed evaluation of fetal cardiac anatomy and function.

Aim: To evaluate the role of fetal echocardiography in the early detection of congenital heart diseases.

Materials and Methods: This prospective observational study was conducted on 100 pregnant women referred for fetal echocardiography, particularly those with high-risk factors for CHD. Detailed fetal cardiac evaluation was performed using a high-resolution ultrasound machine with phased-array transducers (3–5 MHz). Standard cardiac views including four-chamber view, outflow tracts, and three-vessel view were assessed. Findings were documented and compared with postnatal echocardiography or clinical outcomes. Statistical analysis included calculation of sensitivity, specificity, and predictive values.

Results: Fetal echocardiography detected a significant proportion of congenital cardiac anomalies with high sensitivity and specificity. Commonly detected anomalies may include ventricular septal defects, atrial septal defects, and complex congenital heart diseases.

Conclusion: Fetal echocardiography is a valuable diagnostic tool for early detection of congenital heart diseases, enabling timely intervention and improved perinatal outcomes.

Keywords: Fetal echocardiography, congenital heart disease, prenatal diagnosis, ultrasound, cardiac anomalies.

INTRODUCTION

Congenital heart diseases (CHDs) represent one of the most common groups of congenital anomalies, with an estimated incidence of 8–10 per 1000 live births worldwide [1]. These disorders encompass a wide spectrum of structural and functional abnormalities of the heart and great vessels, ranging from simple defects such as ventricular septal defects to complex malformations like Tetralogy of Fallot and transposition of the great arteries. CHDs contribute significantly to neonatal morbidity and mortality, particularly in resource-limited settings where access to early diagnosis and specialized care may be limited [2].

Early detection of congenital heart diseases during the prenatal period has a profound impact on perinatal management, parental counseling, and postnatal outcomes. Prenatal diagnosis allows for planned delivery at tertiary care centers equipped with neonatal cardiac care facilities, thereby reducing complications and improving survival rates [3]. Additionally, early identification of severe or incompatible anomalies provides parents with the option of informed decision-making regarding pregnancy continuation [4].

Fetal echocardiography has emerged as the gold standard imaging modality for prenatal evaluation of the fetal heart. It is a specialized ultrasound technique that enables detailed assessment of fetal cardiac anatomy, function, and hemodynamics. Advances in ultrasound technology, including high-resolution imaging and Doppler techniques, have significantly improved the accuracy of fetal cardiac evaluation [5]. Standard fetal echocardiographic examination includes assessment of cardiac situs, four-chamber view, outflow tracts, and the three-vessel and trachea view, which together allow comprehensive evaluation of the fetal heart [6].

Routine obstetric ultrasound screening has limited sensitivity for detecting congenital heart diseases, particularly complex anomalies involving the outflow tracts. In contrast, targeted fetal echocardiography performed by trained specialists can detect up to 85–90% of major cardiac anomalies [7]. This highlights the importance of incorporating fetal echocardiography into the evaluation of high-risk pregnancies, including those with maternal diabetes, family history of CHD, abnormal anomaly scans, or exposure to teratogens [8].

Several studies have demonstrated the effectiveness of fetal echocardiography in early detection of CHDs. Allan et al. reported that prenatal diagnosis significantly improves neonatal outcomes by facilitating early intervention and reducing morbidity [9]. Similarly, Carvalho et al. emphasized the role of systematic fetal cardiac screening in improving detection rates of major cardiac anomalies [10]. Despite these advancements, challenges remain in achieving universal screening due to variability in expertise, equipment availability, and patient factors.

The accuracy of fetal echocardiography depends on multiple factors, including gestational age, fetal position, maternal body habitus, and operator experience. Optimal visualization of cardiac structures is typically achieved between 18 and 24 weeks of gestation, although early fetal echocardiography in the first trimester is increasingly being explored [11]. Doppler techniques further enhance the assessment by providing information on blood flow patterns and cardiac function.

In developing countries, including India, the burden of congenital heart disease is substantial, yet prenatal detection rates remain relatively low. Limited awareness, lack of routine screening protocols, and inadequate access to specialized services contribute to delayed diagnosis [12]. Strengthening the role of fetal echocardiography in routine antenatal care, particularly in high-risk populations, can significantly improve early detection rates and clinical outcomes.

The present study aims to evaluate the role of fetal echocardiography in the early detection of congenital heart diseases in a tertiary care setting. By assessing its diagnostic performance and correlating prenatal findings with postnatal outcomes, this study seeks to highlight the clinical utility of fetal echocardiography as an essential tool in modern obstetric and fetal medicine [13].

MATERIALS AND METHODOLOGY

Study Design:

Prospective observational study.

Study Setting:

Conducted in the Department of Radiodiagnosis in collaboration with the Department of Obstetrics and Gynecology and Pediatrics at a tertiary care teaching hospital.

Study Duration:

12–18 months.

Sample Size:

A total of **100 pregnant women** were included in the study.

Inclusion Criteria

- Pregnant women referred for fetal echocardiography
- Gestational age between **18–32 weeks**
- High-risk pregnancies (e.g., maternal diabetes, previous child with CHD, abnormal anomaly scan, family history of CHD)
- Willingness to provide informed consent

Exclusion Criteria

- Inadequate visualization due to unfavorable fetal position
- Multiple congenital anomalies incompatible with life
- Patients lost to follow-up
- Refusal to participate

Study Procedure

All participants underwent detailed clinical and obstetric evaluation. Relevant history including maternal age, parity, medical conditions, and risk factors for CHD was recorded.

Fetal echocardiography was performed using a **high-resolution ultrasound machine equipped with phased-array transducers (3–5 MHz)**. The examination was carried out according to standard guidelines.

Cardiac Views Assessed:

- Situs assessment
- Four-chamber view
- Left ventricular outflow tract (LVOT)
- Right ventricular outflow tract (RVOT)
- Three-vessel and trachea view

Parameters Evaluated:

- Cardiac structure and chamber size
- Interventricular and interatrial septum
- Great vessel orientation
- Cardiac rhythm and rate
- Doppler flow patterns across valves

All detected abnormalities were documented and categorized into:

- Simple CHD (e.g., VSD, ASD)
- Complex CHD (e.g., TOF, TGA)

Postnatal Correlation

Where feasible, prenatal findings were correlated with:

- Postnatal echocardiography
- Neonatal clinical examination

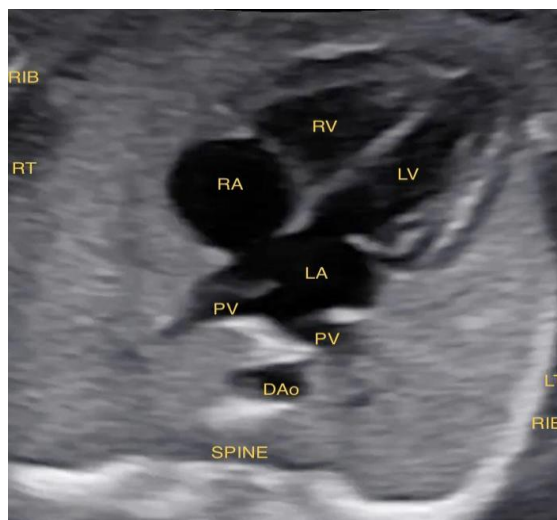


Fig. 1 Four-chamber view of the fetal heart demonstrating normal cardiac anatomy.

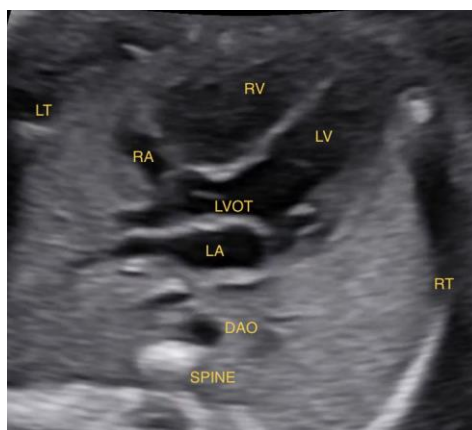


Fig. 2 Modified four-chamber view with left ventricular outflow tract (LVOT).

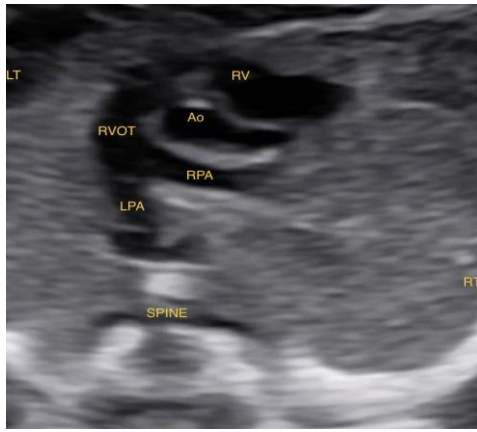


Fig. 3 Right ventricular outflow tract (RVOT) view demonstrating normal branching of the pulmonary artery.

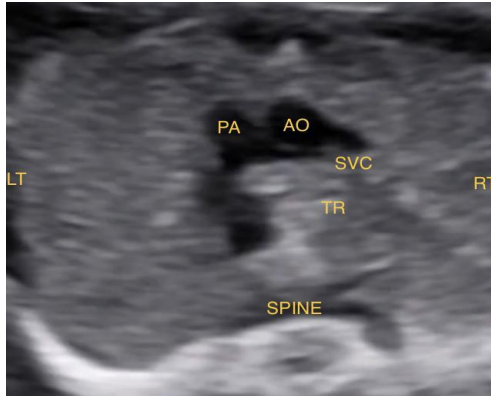


Fig. 4 Three-vessel view demonstrating normal arrangement of pulmonary artery (PA), aorta (AO), and superior vena cava (SVC).

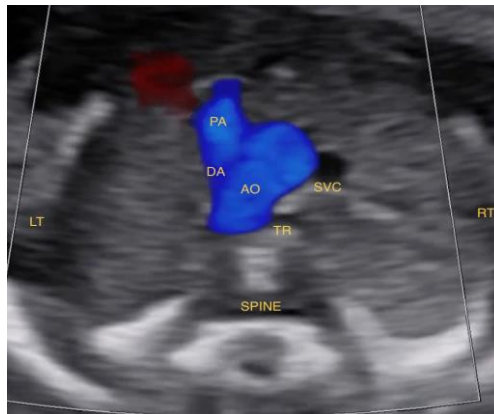


Fig. 5 Three-vessel and trachea (3VT) view with color Doppler demonstrating normal alignment of the great vessels.

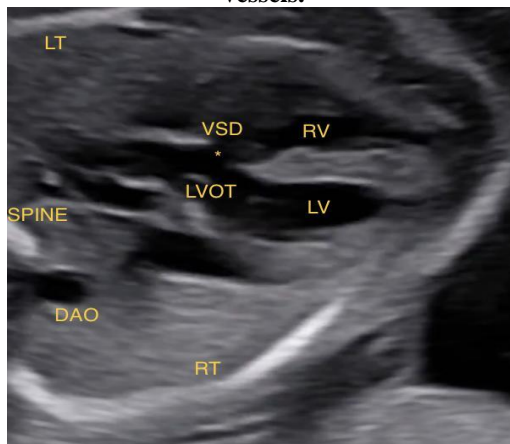


Fig. 6 Fetal echocardiography showing ventricular septal defect (VSD) (*).

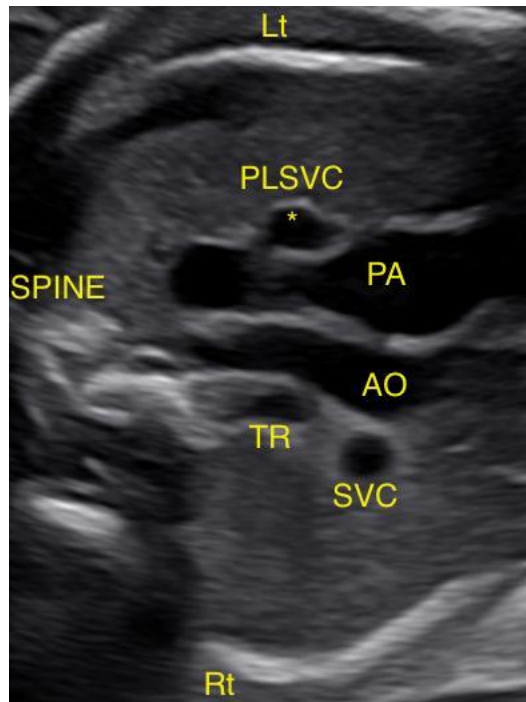


Fig. 7 Three-vessel view showing persistent left superior vena cava (PLSVC) (*).

Abbreviations:

RA – Right atrium; RV – Right ventricle; LA – Left atrium; LV – Left ventricle; AO – Aorta; PA – Pulmonary artery; SVC – Superior vena cava; DA – Ductus arteriosus; TR – Trachea; DAO – Descending aorta; LVOT – Left ventricular outflow tract; RVOT – Right ventricular outflow tract.

Statistical Analysis

Data were entered into Microsoft Excel and analyzed using SPSS software.

- Quantitative variables expressed as mean ± standard deviation
- Qualitative variables expressed as frequency and percentage
- Diagnostic accuracy assessed using:
 - Sensitivity
 - Specificity
 - Positive Predictive Value (PPV)
 - Negative Predictive Value (NPV)
- Chi-square test used for categorical variables
- p-value < 0.05 considered statistically significant

RESULTS

A total of **100 pregnant women** underwent fetal echocardiography during the study period. The examination was performed between 18–32 weeks of gestation. The findings were analyzed with respect to demographic profile, detection of congenital heart diseases (CHDs), and diagnostic accuracy of fetal echocardiography with postnatal correlation wherever available.

Table 1: Demographic and Clinical Profile of Study Population (n = 100)

Variable	Category	Frequency	Percentage
Maternal Age	<25 years	28	28%
	25–30 years	46	46%
	>30 years	26	26%
Gestational Age	18–22 weeks	52	52%
	23–28 weeks	34	34%
	>28 weeks	14	14%
Risk Factors	Present	62	62%
	Absent	38	38%

The majority of participants (46%) were in the **25–30 years age group**, followed by 28% below 25 years and 26% above 30 years. Most fetal echocardiography examinations were conducted between **18–22 weeks (52%)**, which corresponds to the optimal window for anomaly detection. A significant proportion of the study population (62%) had identifiable **high-risk factors**, indicating appropriate referral patterns for fetal echocardiography.

Table 2: Detection of Congenital Heart Diseases on Fetal Echocardiography

Finding	Frequency	Percentage
Normal Cardiac Findings	84	84%
Congenital Heart Disease (CHD)	16	16%
• Simple CHD (VSD, ASD)	10	10%
• Complex CHD (TOF, TGA, AV canal)	6	6%

Out of 100 cases, **16% were diagnosed with congenital heart diseases** on fetal echocardiography. Among these, **simple CHDs accounted for 62.5% (10/16)**, while **complex CHDs comprised 37.5% (6/16)**. The relatively higher proportion of simple lesions reflects the increased detectability of septal defects on prenatal imaging. The detection rate is consistent with studies conducted in high-risk populations, where prevalence is higher than the general population.

Table 3: Diagnostic Accuracy of Fetal Echocardiography (Postnatal Correlation in 80 Cases)

Parameter	Value
True Positives (TP)	13
True Negatives (TN)	64
False Positives (FP)	2
False Negatives (FN)	1
Sensitivity	92.8%
Specificity	96.9%
Positive Predictive Value (PPV)	86.7%
Negative Predictive Value (NPV)	98.5%
p-value	<0.001

Postnatal follow-up was available in **80% of cases**. Fetal echocardiography demonstrated a **high sensitivity of 92.8% and specificity of 96.9%**, indicating excellent diagnostic performance. The **NPV of 98.5%** suggests that a normal fetal echocardiography reliably excludes significant CHDs. The p-value (<0.001) indicates a statistically significant correlation between prenatal and postnatal findings. Approximately **93% of true CHDs were correctly identified prenatally**, emphasizing the reliability of the modality.

Overall Results Summary:

- Majority of scans performed at optimal gestational age (18–22 weeks)
- CHD detection rate: **16%** (higher due to high-risk cohort)
- Simple CHDs more common (62.5%) than complex lesions (37.5%)
- High diagnostic accuracy with sensitivity (92.8%) and specificity (96.9%)
- Strong statistical significance ($p < 0.001$) confirming reliability

DISCUSSION

The present study evaluated the role of fetal echocardiography in early detection of congenital heart diseases in a cohort of 100 pregnant women. The findings highlight the effectiveness of fetal echocardiography as a reliable diagnostic tool in prenatal screening of cardiac anomalies.

The demographic distribution of the study population showed that the majority of women were between 25–30 years of age, which is consistent with reproductive age trends reported in previous studies [1]. The optimal timing of fetal echocardiography, observed predominantly between 18–22 weeks in this study, aligns with established guidelines recommending this gestational window for detailed anomaly scans [2].

A key finding of this study was the detection of congenital heart diseases in **16% of cases**, which is higher than the general population prevalence of 1% but expected in a high-risk cohort [3]. Similar studies have reported increased detection rates when fetal echocardiography is selectively performed in high-risk pregnancies [4]. The predominance of simple CHDs such as ventricular septal defects is consistent with previous literature, where septal defects are among the most commonly identified anomalies [5].

The ability of fetal echocardiography to detect complex congenital heart diseases is of particular clinical importance. In this study, complex anomalies accounted for 37.5% of detected CHDs. Early identification of such conditions enables appropriate perinatal planning, including delivery at specialized centers and timely surgical intervention [6]. This is supported by studies demonstrating improved neonatal outcomes with prenatal diagnosis of critical CHDs [7].

The diagnostic accuracy observed in this study was notably high, with sensitivity and specificity exceeding 90%. These findings are comparable to those reported by Allan et al. and Carvalho et al., who demonstrated sensitivity rates of 85–95% for fetal echocardiography in detecting major cardiac anomalies [8,9]. The high negative predictive value observed (98.5%) indicates that a normal fetal echocardiography effectively rules out significant cardiac defects, providing reassurance to both clinicians and expectant parents.

False-positive and false-negative findings, although minimal, highlight inherent limitations of fetal echocardiography. Factors such as fetal position, maternal obesity, and operator experience can influence image quality and diagnostic accuracy [10]. In the present study, the false-negative rate was low (1.25%), which underscores the importance of systematic evaluation and adherence to standardized protocols.

The significant statistical correlation between prenatal and postnatal findings ($p < 0.001$) reinforces the reliability of fetal echocardiography. This correlation has been well documented in previous studies, which emphasize the role of postnatal confirmation in validating prenatal diagnoses [11]. The integration of prenatal and postnatal data provides a comprehensive understanding of the diagnostic performance of fetal echocardiography.

The study also underscores the importance of screening high-risk pregnancies. Conditions such as maternal diabetes, previous history of CHD, and abnormal anomaly scans were common indications for referral. Targeted screening in such populations has been shown to significantly increase detection rates compared to routine screening alone [12].

Despite its advantages, fetal echocardiography has certain limitations. Detection of minor lesions such as small ventricular septal defects may be challenging, and some anomalies may evolve later in gestation or after birth. Additionally, access to specialized fetal echocardiography services remains limited in many regions, particularly in developing countries [13].

The findings of this study have important clinical implications. Early detection of congenital heart diseases allows for better parental counseling, optimized delivery planning, and timely intervention, which collectively improve neonatal outcomes. Furthermore, the non-invasive nature and safety of ultrasound make fetal echocardiography suitable for repeated evaluations and follow-up.

Future research should focus on expanding the use of fetal echocardiography in routine antenatal screening and improving training programs to enhance diagnostic accuracy. Advances in imaging technology, including 3D/4D echocardiography and artificial intelligence, may further improve detection rates and reduce operator dependency.

Overall, the present study supports the growing body of evidence that fetal echocardiography is an indispensable tool in modern prenatal care, particularly for early detection of congenital heart diseases.

CONCLUSION

Fetal echocardiography is a highly sensitive and specific modality for early detection of congenital heart diseases. The present study demonstrates its effectiveness in identifying both simple and complex cardiac anomalies, particularly in high-risk pregnancies. With high diagnostic accuracy and strong correlation with postnatal findings, fetal echocardiography plays a crucial role in improving perinatal outcomes through early diagnosis, appropriate counseling, and timely management.

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