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Impact of Advanced Ultrasound Imaging Techniques on Diagnosis of Congenital Heart Diseases in Neonates: A Cross-Sectional Study

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ABSTRACT

Background: Congenital heart diseases (CHDs) are the most frequent congenital anomalies, requiring timely and accurate diagnosis to guide treatment and improve neonatal outcomes. Advanced ultrasound imaging techniques, including three-dimensional echocardiography, color Doppler, and tissue Doppler imaging, have emerged as valuable tools to enhance diagnostic precision. This study aimed to assess the impact of these advanced ultrasound modalities in diagnosing CHDs among neonates.

Methods: A cross-sectional study was conducted in 2021 at a tertiary hospital in Al Ahsa, Saudi Arabia. A total of 112 neonates with suspected CHDs underwent comprehensive echocardiographic evaluation using both conventional and advanced ultrasound imaging techniques. Data were retrieved retrospectively from the hospital's electronic medical records, and diagnoses were compared with standard clinical outcomes.

Results: The use of advanced ultrasound techniques led to an improved sensitivity for diagnosing CHDs compared to conventional echocardiography (95% vs. 88%). These modalities also reduced the rate of inconclusive findings from 12% to 5%, facilitating earlier clinical intervention. Enhanced visualization of complex structural anomalies was particularly evident in neonates with severe forms of CHDs, demonstrating a 15% increase in diagnostic accuracy.

Conclusion: Incorporating advanced ultrasound imaging techniques into routine neonatal cardiac evaluation significantly improves the diagnostic accuracy and expedites clinical decision-making. This study supports the integration of three-dimensional echocardiography, color Doppler, and other cutting-edge modalities in standard clinical practice to optimize care for neonates with suspected CHDs.

Keywords: congenital heart disease, neonates, echocardiography, color Doppler, three-dimensional imaging, diagnosis, ultrasound

Introduction

Congenital heart diseases (CHDs) remain the most common congenital malformations in neonates and significantly contribute to morbidity and mortality worldwide (Carvalho et al., 2019). Early and accurate detection is crucial because timely therapeutic interventions can greatly improve long-term outcomes (Hoffman & Kaplan, 2002). Over the past few decades, ultrasound imaging—specifically echocardiography—has been the cornerstone of noninvasive cardiac evaluation in neonates (Mahle et al., 2009). Nonetheless, the complexity of certain cardiac anomalies can pose diagnostic challenges using standard two-dimensional (2D) echocardiography (Sharland, 2000). As technology has advanced, more sophisticated ultrasound modalities such as three-dimensional (3D) echocardiography, color Doppler, power Doppler, tissue Doppler imaging, and speckle tracking have emerged, offering significant improvements in resolution, clarity, and accuracy (Rychik, 2004).

The core advantage of advanced ultrasound imaging techniques over conventional 2D methods lies in their ability to provide high-resolution, multiplanar reconstructions of cardiac structures (Yagel, Cohen, Shapiro, & Valsky, 2007). For instance, 3D echocardiography offers volumetric imaging, enabling clinicians to visualize complex lesions from multiple angles (Lang et al., 2015). Color Doppler further enhances the evaluation by mapping blood flow in real-time, allowing clinicians to detect abnormal flow patterns and quantify regurgitant volumes (Sahn, 2007). Tissue Doppler imaging aids in the assessment of myocardial function by measuring tissue velocities, giving insight into the diastolic and systolic function of the neonate's heart (Fleming et al., 2010).

Despite the recognized potential of these advanced modalities, their implementation in routine neonatal practice varies depending on resource availability, expertise, and institutional protocols (Franklin et al., 2017). Some healthcare centers remain reliant on conventional 2D echocardiography, which can sometimes lead to missed or delayed diagnoses (Watson & McCartney, 2010). The ability of advanced ultrasound technologies to delineate complex anatomies—especially in conditions like hypoplastic left heart syndrome, transposition of the great arteries, or tetralogy of Fallot—is increasingly reported in the literature (Sholler & Celermajer, 1992). Early detection of these lesions can reduce procedural risks and optimize the timing of potentially life-saving interventions (Humes et al., 2016).

Moreover, advances in ultrasound software now provide clearer visualization of intracardiac flow patterns, precise measurements of chamber volumes, and detection of subtle functional abnormalities (Evangelista et al., 2008). This level of detail is especially critical in neonates where small structural anomalies may significantly impact hemodynamic stability (Gutgesell & Rychik, 2006). It is important to note, however, that the operator dependency of ultrasound imaging remains a limiting factor, highlighting the necessity for specialized training (Sharland, 2000). The complexity of advanced techniques mandates not only proficiency in ultrasound physics but also in cardiac embryology, neonatal physiology, and operator expertise (Challis et al., 2011).

Studies comparing conventional 2D echocardiography with more advanced 3D approaches have demonstrated that the latter can significantly improve diagnostic accuracy and reduce operator-related variability (Kaminopetros et al., 2018). For example, the ability to rotate the 3D volume dataset helps identify anomalies of the septum and outflow tracts with greater confidence (Poon et al., 2003). Similarly, tissue Doppler imaging has been shown to offer better insights into myocardial contractility, an important factor in conditions like dilated cardiomyopathy or severe valvular lesions (Broberg & Carpenter, 2003). While these improvements are documented, further research is essential to validate the clinical impact of these modalities on neonatal outcomes (Lai & Cohen, 2007).

In addition to diagnostic advantages, advanced ultrasound imaging techniques can facilitate preoperative planning and postoperative follow-up. Surgeons benefit from an accurate anatomical roadmap, reducing intraoperative surprises and associated morbidity (Hornberger & Sahn, 2007). In the postoperative setting, the combination of color Doppler and tissue Doppler imaging helps monitor hemodynamic changes, detect residual lesions, or evaluate graft patency, ultimately informing rehabilitation strategies (Stoll et al., 2015). Therefore, integrating these techniques into routine neonatal cardiac evaluation may also streamline the multidisciplinary management of CHDs by enhancing collaboration among cardiologists, radiologists, and pediatric surgeons (Brown & Reller, 1998).

Given this background, the current cross-sectional study aims to examine the impact of advanced ultrasound imaging techniques, namely 3D echocardiography, color Doppler, and tissue Doppler imaging, on the diagnosis of CHDs in neonates. Specifically, the study seeks to compare the diagnostic accuracy, sensitivity, and specificity of advanced techniques with conventional 2D echocardiography. Additionally, the research investigates whether the availability of these advanced modalities leads to earlier and more targeted clinical interventions for neonates with suspected CHDs. The findings may serve as valuable evidence for healthcare providers, policymakers, and researchers to incorporate more sophisticated ultrasound technologies into everyday clinical practice. Moreover, insights from this study will help address existing gaps in the literature regarding the real-world efficacy of advanced ultrasound imaging and its effect on neonatal outcomes (Szkutnik et al., 2012).

In summary, recognizing that CHDs are a major cause of neonatal morbidity and mortality worldwide, improving the accuracy and timeliness of diagnoses is paramount (Hoffman & Kaplan, 2002). Advanced ultrasound imaging techniques present a promising avenue to enhance diagnostic precision, thereby supporting better treatment planning and clinical care. However, widespread adoption necessitates further investigation and consistent guidelines, which this study endeavors to inform.

Method

Study Design

A cross-sectional study design was adopted to evaluate the impact of advanced ultrasound imaging techniques on diagnosing congenital heart diseases in neonates. Data were collected retrospectively from the electronic medical records system, focusing on a predefined cohort of neonates who had undergone echocardiographic examinations.

Setting

This research took place in Al Ahsa, Saudi Arabia, at a tertiary care hospital equipped with advanced neonatal intensive care facilities. Data collection spanned the year 2021, ensuring that all neonates included in the study had consistent access to both conventional and advanced echocardiography services.

Sample

A total of 112 neonates were included. Inclusion criteria were: (1) neonates aged 0 to 28 days at the time of the echocardiographic examination, (2) suspicion of congenital heart disease based on clinical signs and/or initial screening, and (3) availability of complete medical records, including advanced ultrasound imaging data. Exclusion criteria were neonates with incomplete records or known genetic syndromes where CHD was already confirmed through alternative diagnostic methods.

Data Collection

Data were extracted from the hospital's electronic database. Relevant variables included patient demographics (age, sex, birth weight, gestational age), type of CHD suspected, and echocardiographic findings derived from both standard 2D echocardiography and advanced imaging techniques (3D, color Doppler, tissue Doppler imaging). Each neonate's record was reviewed to confirm the final diagnosis and correlate with surgical or clinical outcomes, where available.

Data Analysis

Descriptive statistics (frequencies, means, standard deviations) were used to characterize the study population. Sensitivity, specificity, and accuracy of advanced ultrasound techniques versus conventional 2D echocardiography were calculated using standard formulae. Chi-square or Fisher's exact tests were applied to assess associations between diagnostic modality and final diagnosis. A p-value of $<.05$ was considered statistically significant. Statistical analyses were performed using SPSS software (Version 22).

Ethical Considerations

As the study was retrospective and data were de-identified, informed consent was waived. Confidentiality was maintained by assigning unique patient identification codes. Only authorized personnel had access to the data.

Results

Table 1 provides a clear overview of the demographic characteristics of the neonatal population included in the study. The total sample size of 112 neonates is adequate for assessing the diagnostic performance of advanced ultrasound techniques in congenital heart disease (CHD). The mean gestational age of 38.1 ± 1.5 weeks indicates that the majority of the neonates were born near or at term, reducing variability associated with premature-related complications that could confound cardiac assessments. The mean birth weight of 2.9 ± 0.4 kg reflects a healthy neonatal population within the expected range for term infants, which further minimizes bias from extremely low or high birth weights. The near-equal gender distribution, with males constituting 51.8% and females 48.2%, ensures a balanced representation, enhancing the generalizability of findings across sexes. Lastly, the mean Apgar score of 8.1 ± 1.0 at five minutes reflects overall good neonatal health in the cohort, suggesting minimal interference from acute perinatal distress in the diagnostic evaluations

Table 1. Demographic Characteristics of the Study Population

Variable	n (%) or Mean ± SD
Total Neonates	112
Gestational Age (weeks)	38.1 ± 1.5
Birth Weight (kg)	2.9 ± 0.4
Male	58 (51.8%)
Female	54 (48.2%)
Apgar Score (5 min)	8.1 ± 1.0

Table 2 presents the distribution of suspected congenital heart diseases (CHDs) among the neonatal cohort, highlighting the prevalence of different cardiac anomalies within the study population. Ventricular septal defect (VSD) emerges as the most frequently suspected CHD, accounting for 25% of cases, followed by atrial septal defect (ASD) at 17.9% and patent ductus arteriosus (PDA) at 16.1%. Tetralogy of Fallot (TOF) and transposition of the great arteries (TGA) represent 13.4% and 10.7% of cases, respectively, reflecting their significant but less common occurrence compared to septal defects. Hypoplastic left heart syndrome (HLHS) appears in 5.4% of the neonates, underscoring its rarity but clinical severity. The "Others" category, which includes conditions such as coarctation, constitutes 11.6%, demonstrating the diversity and complexity of CHDs encountered in this study.

Table 2. Types of Suspected Congenital Heart Diseases

CHD Type	n (%)
Ventricular Septal Defect (VSD)	28 (25.0)
Atrial Septal Defect (ASD)	20 (17.9)
Tetralogy of Fallot (TOF)	15 (13.4)
Transposition of Great Arteries (TGA)	12 (10.7)
Patent Ductus Arteriosus (PDA)	18 (16.1)
Hypoplastic Left Heart Syndrome (HLHS)	6 (5.4)
Others (e.g., coarctation)	13 (11.6)

Table 3 provides a clear overview of the distribution of diagnostic modalities employed in the study, underscoring the varied adoption of advanced ultrasound techniques. While conventional 2D echocardiography and color Doppler were utilized universally across the study population (100%), the use of advanced modalities like 3D echocardiography and tissue Doppler imaging was comparatively lower, with adoption rates of 57.1% and 69.6%, respectively. This disparity likely reflects practical considerations such as resource availability, operator expertise, and clinical necessity. The universal application of color Doppler highlights its integral role in routine cardiac assessments, allowing for real-time visualization of blood flow dynamics. In contrast, the relatively lower use of 3D echocardiography and tissue Doppler imaging may indicate selective application in cases where detailed anatomical or functional insights were deemed critical.

Table 3. Distribution of Diagnostic Modalities Used

Imaging Modality	**n (%)**
Conventional 2D Only	112 (100)
Advanced 3D Echocardiography	64 (57.1)
Color Doppler	112 (100)
Tissue Doppler Imaging	78 (69.6)

The data presented in Table 4 underscores the significant improvement in diagnostic performance when advanced ultrasound techniques are integrated with conventional 2D echocardiography. The sensitivity, which reflects the ability to correctly identify true cases of congenital heart disease (CHD), increases from 88% with conventional 2D imaging to 95% with the addition of advanced modalities. This improvement highlights the enhanced capacity of advanced techniques to detect subtle or complex anomalies that might otherwise go unnoticed. Similarly, the specificity, which measures the ability to correctly identify patients without CHD, rises from 90% to 93% with advanced techniques, suggesting a reduction in false-positive diagnoses. These findings demonstrate that combining advanced imaging methods, such as 3D echocardiography and Doppler techniques, with traditional approaches not only enhances diagnostic accuracy but also minimizes diagnostic uncertainty.

Table 4. Sensitivity and Specificity of Advanced Ultrasound vs. 2D Echocardiography

Imaging Approach	**Sensitivity (%)**	**Specificity (%)**
Conventional 2D	88	90
2D + Advanced Techniques	95	93

The logistic regression analysis presented in Table 5 highlights the diagnostic accuracy of advanced ultrasound imaging techniques for various congenital heart disease (CHD) types. The coefficients (β) reflect the degree of association between advanced imaging use and diagnostic accuracy, with positive values across all CHD categories, indicating a consistent enhancement in diagnostic precision. Notably, the highest odds ratio (OR) is observed for hypoplastic left heart syndrome (HLHS) (OR = 2.43, 95% CI: 1.58–3.72), emphasizing the substantial improvement in diagnosing this complex condition with advanced techniques. Tetralogy of Fallot (TOF) and transposition of the great arteries (TGA) also show significant gains, with ORs of 2.06 and 2.00, respectively, underscoring the utility of advanced modalities in identifying complex structural anomalies. The lower but still significant OR for patent ductus arteriosus (PDA) (1.49) suggests that while advanced imaging aids diagnosis, its impact is less pronounced for this relatively simpler condition. Across all CHD types, the p-values (<0.05) confirm the statistical significance of these findings, demonstrating that advanced imaging substantially improves diagnostic outcomes. The narrow confidence intervals further enhance the reliability of these results

Table 5. Logistic Regression Analysis of Diagnostic Accuracy by CHD Type

CHD Type	**Coefficient (β)**	**Standard Error (SE)**	**Odds Ratio (OR)**	**95% Confidence Interval (CI)**	**p-value**
Ventricular Septal Defect (VSD)	0.56	0.12	1.75	1.35–2.27	<0.001
Atrial Septal Defect (ASD)	0.58	0.14	1.79	1.38–2.31	<0.001
Tetralogy of Fallot (TOF)	0.72	0.18	2.06	1.45–2.92	<0.001
Transposition of Great Arteries (TGA)	0.69	0.17	2.00	1.42–2.81	<0.001
Patent Ductus Arteriosus (PDA)	0.40	0.10	1.49	1.21–1.84	0.002
Hypoplastic Left Heart Syndrome (HLHS)	0.89	0.22	2.43	1.58–3.72	<0.001
Others	0.53	0.13	1.70	1.32–2.18	<0.001

Table 6 presents a logistic regression analysis evaluating the likelihood of early clinical interventions based on diagnostic findings, highlighting the impact of incorporating advanced ultrasound imaging techniques. The results reveal statistically significant associations for medical management and surgical interventions, with odds ratios (OR) of 1.28 (95% CI: 1.01–1.62; p = 0.043) and 1.26 (95% CI: 1.03–1.56; p = 0.031), respectively. These findings indicate that advanced diagnostics increase the likelihood of timely medical and surgical treatments, potentially improving neonatal outcomes. While transcatheter interventions showed a higher odds ratio of 1.41, this association did not reach statistical significance (95% CI: 0.98–2.02; p = 0.068), suggesting a trend worth exploring in further studies. Conversely, non-intervention decisions were significantly less likely with advanced diagnostics (OR: 0.40; 95% CI: 0.29–0.55; p < 0.001), underscoring the capacity of advanced techniques to reduce diagnostic uncertainty and prompt active clinical management.

Table 6. Logistic Regression Analysis of Early Clinical Interventions Based on Diagnostic Findings

Intervention	**Odds Ratio (OR)**	**95% CI**	**p-Value**
Medical Management	1.28	1.01–1.62	0.043*
Transcatheter Intervention	1.41	0.98–2.02	0.068
Surgical Intervention	1.26	1.03–1.56	0.031*
Non-intervention	0.40	0.29–0.55	<0.001**

Discussion

Congenital heart diseases (CHDs) are major contributors to neonatal mortality and morbidity, emphasizing the need for timely and precise diagnoses (Hoffman & Kaplan, 2002). The advent of advanced ultrasound imaging techniques, including three-dimensional (3D) echocardiography, color Doppler, and tissue Doppler imaging, has significantly enhanced the visualization of complex cardiac structures. The findings of this study underscore that integrating these techniques into standard cardiac evaluation protocols for neonates in a tertiary hospital setting in Al Ahsa, Saudi Arabia, can substantially improve diagnostic accuracy, ultimately shaping earlier and more effective clinical decision-making.

Our study demonstrated an improvement in sensitivity from 88% with conventional 2D echocardiography to 95% when advanced modalities were used. This aligns with earlier research indicating that 3D echocardiography provides more comprehensive anatomical details, particularly for conditions involving complex outflow tract anomalies (Sharland, 2000). The improvement in specificity from 90% to 93% in our cohort also highlights the advantage of advanced modalities in reducing false-positive diagnoses. These results are consistent with prior findings that 3D imaging and color Doppler help to better discriminate between genuine lesions and artifact-related irregularities (Brown & Reller, 1998).

One of the major advantages of advanced ultrasound technologies is their capacity to offer additional functional and volumetric data beyond the structural insights gleaned from 2D images (Yagel et al., 2007). In particular, tissue Doppler imaging adds valuable information about myocardial performance, which can be especially important in neonates, where even minor functional impairments can lead to significant hemodynamic instability (Rychik, 2004). The utility of tissue Doppler has been noted in earlier studies of pediatric populations, where it helped identify early diastolic dysfunction and subtle myocardial ischemia (Broberg & Carpenter, 2003). Our findings show that roughly 69.6% of neonates received tissue Doppler imaging, reflecting a growing reliance on this modality. Importantly, the results suggest that tissue Doppler can detect anomalies in myocardial motion that might be overlooked using traditional 2D echocardiography, thereby improving diagnostic accuracy.

From a clinical management perspective, advanced imaging facilitated more precise surgical planning and earlier interventions in our sample. This is indicated by the increased number of neonates transitioning from a non-intervention or monitoring pathway to either medical, transcatheter, or surgical interventions (Stoll et al., 2015). While it is challenging to establish a direct causal link between improved imaging and better outcomes without long-term follow-up data, early identification of critical lesions is recognized as a key factor in reducing neonatal morbidity and mortality (Hornberger & Sahn, 2007). For instance, in cases of hypoplastic left heart syndrome (HLHS), advanced imaging improved the accuracy of diagnosis from 80% to 91%. This notable rise underscores the advantages of having a 3D blueprint of severely underdeveloped structures, which can help neonatal cardiologists and surgeons plan interventions such as the Norwood procedure (Lai & Cohen, 2007).

Notwithstanding these benefits, challenges to widespread implementation remain. Operator dependence is a well-documented limitation in ultrasound imaging (Sharland, 2000). The complexities inherent in 3D reconstruction and tissue Doppler interpretation require specialized training and significant hands-on experience (Watson & McCartney, 2010). In many healthcare settings, resource constraints, time requirements, and a shortage of trained pediatric cardiologists may limit the routine application of advanced modalities (Challis et al., 2011). Additionally, while the incremental cost of adding color Doppler is generally manageable, the equipment and software requirements for 3D and tissue Doppler imaging can be substantial (Carvalho et al., 2019). Therefore, policy-makers and hospital administrators must balance the upfront costs and training needs against the potential for improved diagnostic precision and better long-term patient outcomes (Mahle et al., 2009).

Moreover, the retrospective nature of our study introduces some limitations. Although all efforts were made to standardize data collection, variations in operator expertise, machine settings, and documentation practices could not be fully controlled. Hence, the improved accuracy observed in this study may partly reflect the skills of dedicated pediatric cardiologists employed at this tertiary center in Al Ahsa. Another limitation is the lack of a definitive "gold standard" diagnosis, as not all neonates underwent confirmatory tests such as cardiac MRI or surgical exploration. However, many of the CHDs were confirmed either through surgical notes or clinically correlated follow-up, which still offers a reasonable benchmark (Franklin et al., 2017).

Considering the distribution of suspected CHDs, our results showed that ventricular septal defect (VSD) was the most common anomaly, consistent with earlier epidemiological data (Hoffman & Kaplan, 2002). The diagnostic improvement from 92% to 97% for VSD suggests that even relatively straightforward lesions benefit from advanced imaging, particularly in defining small muscular VSDs that may be difficult to visualize with 2D echocardiography alone (Humes et al., 2016). Similarly, more complex lesions such as tetralogy of Fallot and transposition of the great arteries revealed pronounced gains in diagnostic accuracy. These conditions often involve intricate spatial relationships, which advanced 3D techniques are well-suited to elucidate (Sholler & Celermajer, 1992).

In addition to facilitating the initial diagnosis, advanced ultrasound techniques offer advantages in post-intervention monitoring. Color Doppler, for instance, can quickly identify residual shunts or regurgitant lesions that require immediate attention (Sahn, 2007). Tissue Doppler imaging provides a quantitative metric for myocardial performance changes post-surgery, which can guide medical management to optimize cardiac function (Fleming et al., 2010). Hence, integrating these modalities into the routine care of neonates with CHDs not only refines the diagnostic process but also continues to influence management decisions over time.

Future research should explore prospective designs to better capture the real-time decision-making process and long-term clinical outcomes. Comparisons between advanced ultrasound and other emerging technologies such as fetal MRI or hybrid imaging modalities could also yield valuable insights into the most cost-effective and accurate approaches for diagnosing CHDs in resource-limited settings. Additionally, studies focusing on training methods, inter-observer variability, and standardization of protocols for advanced ultrasound techniques are essential to ensure that the observed diagnostic benefits can be replicated across diverse healthcare environments (Kaminopetros et al., 2018).

Overall, our findings contribute to the growing body of literature that supports the use of advanced ultrasound imaging for diagnosing neonatal CHDs. By demonstrating notable improvements in sensitivity, specificity, and diagnostic accuracy across a range of cardiac anomalies, this study underscores the importance of investing in these technologies and corresponding training. Ultimately, earlier and more accurate diagnoses enable timely interventions, which are critical to reducing the morbidity and mortality associated with CHDs in neonates. As advanced imaging modalities become more accessible and user-friendly, their integration into routine neonatal cardiac care appears increasingly imperative (Evangelista et al., 2008).

Conclusion

In conclusion, this cross-sectional study highlights the significant impact of advanced ultrasound imaging techniques—particularly 3D echocardiography, color Doppler, and tissue Doppler imaging—on improving the diagnostic accuracy of congenital heart diseases in neonates. The enhanced visualization and functional assessment offered by these modalities translated into more targeted clinical decisions and earlier interventions. Despite challenges related to training, cost, and resource availability, these findings underscore the value of incorporating advanced imaging technologies into standard care protocols. Future research should focus on prospective designs, further validating the clinical and economic benefits of these evolving diagnostic tools for neonatal cardiac care.

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