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Article

Capabilities Of Modern Echocardiography Methods in the Diagnosis of Congenital Heart Anomalies

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Abstract: *Congenital heart defects (CHD) represent the most common structural anomalies in newborns and remain a leading cause of infant morbidity and mortality worldwide. Early and accurate diagnosis is essential for timely intervention and improved long-term outcomes. This review examines the diagnostic capabilities of modern echocardiographic methods — including transthoracic, transesophageal, fetal, three-dimensional (3D), and speckle-tracking echocardiography (STE) — in the detection and management of CHD. An analysis of recent domestic and international scientific publications was conducted to highlight the informational value, advantages, and limitations of each modality. The review demonstrates that the integration of advanced echocardiographic techniques substantially improves prenatal and postnatal CHD detection rates, enhances myocardial function assessment, and supports surgical planning.*

Keywords: *congenital heart anomalies, congenital heart defects, Doppler ultrasonography, cardiology, prenatal diagnosis, ultrasound diagnostics, echocardiography, speckle-tracking, three-dimensional echocardiography.*

1. INTRODUCTION

Congenital heart defects (CHD) are among the most prevalent structural birth anomalies, occurring in approximately 8–10 per 1,000 live births globally [1]. Despite significant advances in pediatric cardiology over recent decades, CHD remain a major contributor to infant morbidity, mortality, and long-term cardiovascular complications [2]. Accurate and timely diagnosis — both prenatally and postnatally — is crucial to guide appropriate clinical management and surgical intervention.

Modern ultrasound imaging methods play an extremely important role in the diagnostic process and are of high value in the comprehensive assessment of pathological cardiac conditions. Due to their high informational value, non-invasiveness, and radiation-free nature, they are widely used in clinical practice for the detection, clarification, and follow-up monitoring of congenital and acquired cardiac diseases [3].

The survival of patients with CHD into adulthood has improved substantially owing to advances in prenatal diagnosis, refined surgical techniques, and the emergence of specialized adult congenital heart disease (ACHD) programs. However, long-term cardiovascular morbidity, including heart failure, persists in this population, with overall mortality from heart failure reported at approximately 4% [4]. This underscores the continuing need for precise, reproducible imaging tools throughout the patient's lifetime.

The primary aim of this review is to systematically evaluate the capabilities of contemporary echocardiographic modalities — transthoracic echocardiography (TTE), transesophageal echocardiography (TEE), fetal echocardiography, three-dimensional echocardiography (3DE), and speckle-tracking echocardiography (STE) — in the diagnosis, functional assessment, and perioperative management of CHD, based on a synthesis of recent literature.

2. METHODS

This paper constitutes a narrative literature review. A systematic search of PubMed/MEDLINE, Scopus, and Google Scholar databases was conducted for publications from 1999 to 2026. The following search terms were used: "echocardiography," "congenital heart defects," "speckle-tracking echocardiography," "fetal echocardiography," "three-dimensional echocardiography," "myocardial strain," and "prenatal diagnosis." Only peer-reviewed original articles, systematic reviews, and expert consensus documents published in English were included. Studies focusing exclusively on acquired heart disease without relevance to CHD were excluded. A total of 15 sources were selected for inclusion based on their relevance to the study objectives and methodological quality.

The identified publications were categorized according to the echocardiographic method described: (1) conventional two-dimensional (2D) echocardiography and Doppler techniques; (2) fetal echocardiography; (3) speckle-tracking echocardiography and myocardial strain analysis; and (4) three-dimensional echocardiography. The findings were synthesized narratively to describe the diagnostic capabilities, clinical applications, and limitations of each modality.

3. RESULTS

3.1. Conventional 2D Echocardiography and Doppler Techniques

Two-dimensional real-time ultrasound (2D ultrasound) has long been the cornerstone tool for both postnatal and fetal cardiac imaging. Continuous improvements in hardware and post-processing software have led to excellent image quality even in the late first trimester [5]. Standard 2D echocardiography with Doppler enables reliable assessment of cardiac anatomy, valve morphology, and hemodynamic parameters such as flow velocities, pressure gradients, and shunt detection across septal defect.

Fetal echocardiography — a comprehensive 2D and Doppler ultrasound examination of the fetal cardiovascular system — allows detailed assessment of cardiac anatomy and functional status during the prenatal period. This method is completely non-invasive, free of ionizing radiation, and can be performed repeatedly for longitudinal follow-up monitoring. It is regarded as a functional analogue of the fetal electrocardiogram, enabling indirect assessment of the electrical and mechanical activity of the fetal heart [6].

Nevertheless, 2D ultrasound retains inherent limitations in spatial resolution for complex three-dimensional cardiac structures, and its diagnostic accuracy is highly operator-dependent. Performing a comprehensive fetal cardiac examination remains challenging for less experienced specialists, as it requires strict adherence to methodology for acquiring sequential transverse and sagittal cardiac views [7]. Incomplete examination steps may lead to reduced detection rates of fetuses with CHD, highlighting the need for standardized protocols and enhanced training.

3.2. Speckle-Tracking Echocardiography and Myocardial Strain Analysis

Speckle-tracking echocardiography (STE) is a relatively recent technique enabling the characterization and quantification of myocardial deformation. By tracking natural acoustic markers (speckles) frame-by-frame, STE measures multiple components of myocardial strain — including longitudinal, circumferential, and radial strain — as well as strain rate, velocity, displacement, rotation, and systolic/diastolic timing [8]. Critically, STE provides hemodynamic information not available from conventional parameters such as left ventricular ejection fraction (LVEF), which may remain normal even in the presence of subclinical myocardial dysfunction [9].

In the adult CHD population, myocardial strain analysis using STE has emerged as a sensitive, non-invasive method for detecting early myocardial dysfunction and supporting clinical decision-making [10]. The number of publications on STE has grown rapidly over the past decade, reflecting its expanding clinical applicability [9]. However, for widespread use, validation across vendor-independent platforms remains necessary.

In pediatric cardiac practice, STE is currently regarded primarily as a research tool, with its routine clinical implementation limited by the lack of vendor-independent normative data across different age groups and cardiac anatomies [11]. Nonetheless, STE has been validated against more complex, time-consuming, and costly techniques and is increasingly valued as a bedside imaging adjunct.

Fetal two-dimensional speckle-tracking echocardiography (2D-STE) represents a novel extension of this technology to the prenatal period. It provides assessment of global longitudinal strain (GLS) and global longitudinal strain rate (GLSR), allowing quantification of longitudinal deformation of the fetal cardiac wall [12]. The identification of fetal myocardial maturation patterns and the early detection of contractile dysfunction using 2D-STE are considered important prerequisites for its further implementation in routine prenatal cardiac assessment [13].

3.3. Three-Dimensional Echocardiography

Three-dimensional echocardiography (3DE) represents a modern and rapidly evolving cardiac imaging modality that enables real-time volumetric visualization of cardiac structures. Major technological advances in ultrasound imaging have ushered in a new era in which 3DE is gradually becoming one of the primary and most informative methods for assessing cardiac anatomy and function [14].

In the context of CHD, 3DE is particularly valuable for preoperative planning, navigation and guidance of catheter-based interventions, and detailed functional assessment of complex cardiac malformations. The excellent acoustic windows available in pediatric patients, combined with the non-invasive and radiation-free nature of the technique, favor its growing adoption in pediatric cardiology [15]. Three- and four-dimensional ultrasound modalities have also demonstrated utility in fetal echocardiography, providing enhanced spatial understanding of complex fetal cardiac anatomy.

Despite these advantages, 3DE remains limited by lower temporal resolution compared with 2D imaging, susceptibility to stitching artifacts in real-time full-volume acquisition, and a steeper learning curve. Current expert consensus documents advocate for its integration as a complementary tool alongside standard 2D echocardiography in the evaluation of CHD.

4. DISCUSSION

The findings of this review confirm that modern echocardiographic methods collectively constitute a comprehensive and powerful diagnostic armamentarium for CHD. Each modality contributes unique and complementary information that, when integrated, substantially enhances the accuracy of structural and functional cardiac assessment across the lifespan — from fetus to adult.

Conventional 2D echocardiography with Doppler remains the diagnostic backbone for initial CHD detection, given its wide availability, low cost, and real-time anatomical visualization. However, its operator dependency and two-dimensional perspective limit its ability to characterize complex spatial relationships within malformed cardiac structures. The supplementation of 2D imaging with 3DE addresses many of these spatial limitations, providing volumetric data that facilitate more accurate anatomical delineation and surgical planning.

STE has transformed the assessment of myocardial mechanics by enabling the quantification of subtle deformation parameters beyond the reach of conventional echocardiography. The clinical importance of early subclinical myocardial dysfunction detection — particularly in patients with surgically corrected CHD — cannot be overstated, as these individuals face a lifelong risk of progressive ventricular impairment. GLS derived from STE has emerged as a reproducible, sensitive marker in this regard.

Fetal echocardiography, particularly when augmented by 2D-STE, represents a pivotal advance in prenatal medicine, enabling not only anatomical diagnosis of CHD but also functional characterization of fetal myocardial physiology. The ability to detect contractile dysfunction prenatally opens new avenues for antenatal counseling and perinatal management planning. However, challenges related to fetal position, acoustic window quality, and the absence of standardized normative fetal strain data across gestational ages remain barriers to routine clinical implementation.

Several important limitations warrant acknowledgment. First, the lack of vendor-independent STE software standardization continues to impede cross-platform comparison and the establishment of universal reference ranges. Second, 3DE is not yet universally accessible, and its use demands specialized training and dedicated acquisition protocols. Third, the evidence base for fetal STE in CHD, while growing, remains largely observational, and prospective longitudinal studies linking prenatal strain parameters to postnatal outcomes are needed. Addressing these gaps through multicenter collaborative research will be essential to transition these technologies from predominantly research applications to validated routine clinical tools.

5. CONCLUSION

Modern echocardiographic technologies — encompassing conventional 2D Doppler imaging, fetal echocardiography, speckle-tracking echocardiography, and three-dimensional echocardiography — collectively provide a powerful, non-invasive, and radiation-free diagnostic framework for the detection, characterization, and longitudinal management of congenital heart defects. The progressive integration of these modalities into clinical practice has measurably improved prenatal CHD detection rates, enabled earlier identification of subclinical myocardial dysfunction, and enhanced preoperative and interventional planning.

Further development and clinical validation of these echocardiographic technologies — particularly vendor-independent STE standardization, expansion of normative fetal strain databases, and broader adoption of 3DE in pediatric and adult CHD settings — represent critical priorities for improving diagnostic quality, reducing diagnostic errors, and optimizing long-term outcomes in this vulnerable patient population.

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