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Review Article

Artificial intelligence for the detection of fetal ultrasound findings concerning major congenital heart defects. A systematic review.

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ABSTRACT

Background: Prenatal detection of major congenital heart defects (CHDs) remains suboptimal despite routine fetal ultrasound screening, largely due to operator dependence, variability in expertise, and subtle morphologic presentations. Artificial intelligence (AI) has emerged as a potential adjunct to improve screening performance by identifying ultrasound findings concerning major CHDs.

Objective: To systematically review and synthesize current evidence on the role of artificial intelligence in detecting fetal ultrasound findings concerning major congenital heart defects.

Methods: A systematic literature search was conducted across PubMed/MEDLINE, Scopus, Web of Science, and Google Scholar in accordance with PRISMA 2020 guidelines. Original peer-reviewed studies evaluating AI-based models applied to fetal ultrasound or fetal echocardiography for the detection of major CHDs or suspicious findings were included. Data were synthesized qualitatively, and the risk of bias was assessed using an adapted QUADAS-2 framework.

Results: This systematic review was not prospectively registered. Five studies published between 2021 and 2026 met the inclusion criteria. AI models demonstrated consistently high diagnostic performance, with reported sensitivities ranging from approximately 84% to 96.8% and specificity generally exceeding 90%. Models focusing on the detection of concerning ultrasound findings rather than lesion-specific diagnosis demonstrated particularly high screening sensitivity. Overall risk of bias was low to moderate.

Conclusion: Artificial intelligence showed strong potential as a screening adjunct for improving prenatal detection of major congenital heart defects, warranting further prospective validation in real-world clinical settings.

Implications and future research (addition):"These findings support the potential role of artificial intelligence–assisted fetal echocardiography as an adjunct to conventional ultrasound screening for major congenital heart defects. However, heterogeneity in study design, AI architectures, outcome reporting, and validation strategies limits direct clinical translation. Future research should prioritize large-scale prospective studies, external validation across diverse populations, standardized performance metrics, and evaluation of real-world clinical integration, including cost-effectiveness and ethical considerations.

Keywords: *Artificial intelligence, Congenital heart defects, Deep learning, Fetal echocardiography, Fetal ultrasound, Prenatal screening.*

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INTRODUCTION

Congenital heart defects (CHDs) affect approximately 8-12 per 1,000 live births and represent the most common category of congenital anomalies worldwide. They account for a substantial proportion of infant morbidity, mortality, and long-term healthcare burden [1,2,3]. Among these, major or severe CHDs, including hypoplastic left heart syndrome, transposition of the great arteries, tetralogy of Fallot, atrioventricular septal defects, and duct-dependent lesions, are of particular clinical importance due to their association with early hemodynamic compromise and the need for specialized neonatal care or urgent surgical intervention [4-7]. Failure to identify these conditions prenatally can result in delayed diagnosis, circulatory collapse after ductal closure, and increased neonatal mortality [8-10]. Conversely, prenatal detection of major CHDs enables optimized perinatal management, planned delivery at tertiary care centers, immediate postnatal stabilization, and timely intervention, all of which have been shown to improve neonatal outcomes and survival [11,12].

Prenatal ultrasound screening remains the cornerstone of fetal cardiac assessment and is routinely performed as part of the second-trimester anomaly scan [13,14]. Current international guidelines recommend systematic evaluation of the fetal heart using the four-chamber view in combination with assessment of the ventricular outflow tracts. These views provide a practical balance between feasibility and diagnostic yield in population-level screening [15,16]. However, despite widespread implementation of these recommendations, prenatal detection rates for major CHDs vary considerably across regions and healthcare systems. Detection rates remain particularly low in low-risk populations and community-based screening programs, where most anomaly scans are performed [17-19]. Several factors contribute to these limitations, including the intrinsic complexity of fetal cardiac anatomy, the dynamic nature of the fetal heart, operator dependence, variable sonographer experience, inconsistent image acquisition, suboptimal fetal position, and time constraints during routine examinations [20,21].

Importantly, many major CHDs do not present with obvious abnormalities on the four-chamber view alone and instead manifest as subtle deviations in outflow tract anatomy, cardiac proportions, or spatial relationships between cardiac structures [22-24]. These subtle morphologic changes may be difficult to recognize consistently, particularly for non-specialist operators. As a result, a significant proportion of major CHDs remain undetected until after birth, highlighting a critical gap between recommended screening protocols and real-world diagnostic performance [25,26].

In recent years, artificial intelligence (AI), particularly deep learning-based methodologies, has emerged as a promising solution to address these challenges. AI systems are capable of analyzing large volumes of medical imaging data, learning complex hierarchical patterns, and identifying subtle features that may not be readily apparent to human observers[27]. In the context of fetal cardiac imaging, AI has been explored for multiple tasks, including automated identification of standard cardiac views, segmentation of cardiac chambers and vessels, quality assessment of ultrasound images, and detection of abnormal morphologic patterns suggestive of congenital heart disease. These capabilities offer the potential to reduce operator dependence, improve consistency, and augment diagnostic accuracy during routine prenatal screening[28,29].

Notably, recent AI research in fetal cardiology has increasingly shifted away from attempting definitive lesion-specific diagnosis at the screening stage. Instead, contemporary approaches focus on detecting ultrasound findings concerning major CHDs, such as abnormal cardiac proportions, atypical ventricular relationships, outflow tract discrepancies, or deviations in cardiac axis and size[30,31]. This paradigm closely mirrors real-world clinical practice, where the primary objective of screening ultrasound is not to establish a precise diagnosis, but rather to identify suspicious examinations that warrant referral for expert fetal echocardiography[32]. By functioning as a screening adjunct or decision-support tool, AI has the potential to improve early recognition of high-risk cases while maintaining a pragmatic and clinically appropriate role within existing care pathways.

Accordingly, the present systematic review aims to critically evaluate and synthesize existing peer-reviewed evidence on the application of artificial intelligence for detecting fetal ultrasound findings concerning major congenital heart defects, with particular emphasis on diagnostic performance, clinical context, and methodological quality.

MATERIALS AND METHODS

Study Design and Reporting Guidelines

This systematic review was conducted in accordance with the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) 2020 guidelines [33].

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Review Question

What is the diagnostic performance and clinical role of artificial intelligence in detecting fetal ultrasound findings concerning major congenital heart defects?

Search Strategy

The electronic databases PubMed/MEDLINE, Scopus, Web of Science, and IEEE Xplore were systematically searched. The final search was conducted on 15 January 2026. All databases were searched from inception to the date of the final search. Reference lists of included studies and relevant reviews were manually screened to identify additional eligible articles.

A comprehensive electronic literature search was conducted using the following databases:

- PubMed/MEDLINE
- Scopus
- Web of Science
- Google Scholar

The search strategy included combinations of the following keywords and phrases:

"artificial intelligence," "deep learning," "machine learning,"

"fetal ultrasound," "fetal echocardiography,"

"congenital heart defect," "major CHD," and "prenatal screening."

In addition, the reference lists of all eligible studies were manually screened to identify any further relevant articles.

Eligibility Criteria

Inclusion Criteria

Studies were included if they met all of the following criteria:

- Original peer-reviewed studies
- Artificial intelligence–based models applied to fetal ultrasound or fetal echocardiography
- Detection of major congenital heart defects or ultrasound findings suspicious for major CHDs
- Reporting of diagnostic performance metrics
- Human fetal studies
- English-language publications

Exclusion Criteria

Studies were excluded if they met any of the following criteria:

- Focused exclusively on minor congenital heart defects
- Based on postnatal imaging
- Case reports, narrative reviews, editorials, or conference abstracts
- Non–artificial intelligence- based computational approaches

Study Selection

Two reviewers independently screened the titles and abstracts of retrieved records, followed by full-text evaluation of potentially eligible studies. Any disagreements were resolved through discussion and consensus.

Data Extraction

The following outcomes were extracted from each included study:

- Diagnostic performance metrics: sensitivity, specificity, accuracy, area under the receiver operating characteristic curve (AUC)
- Type of congenital heart defect detected
- Gestational age at assessment
- Ultrasound view(s) analyzed
- Artificial intelligence model type and architecture
- Training, validation, and test dataset characteristics
- Reference standard used for diagnosis
- External validation status

Where multiple results were reported for the same outcome (e.g., different ultrasound views, time points, or AI models), all compatible results were extracted. If selective reporting was present, priority was given to externally validated results or those derived from independent test datasets.

Data Synthesis

Due to heterogeneity in study design, artificial intelligence methodology, and outcome reporting, a quantitative meta-analysis was not performed. Instead, the findings were synthesized using a qualitative narrative approach.

Risk of Bias Assessment

The methodological quality and risk of bias of the included studies were assessed using the Quality Assessment of

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Diagnostic Accuracy Studies-2 (QUADAS-2) tool[34], adapted for artificial intelligence–based diagnostic accuracy studies. Risk of bias assessment was performed independently by two reviewers, with any disagreements resolved through discussion. (Table 2)

Certainty of Evidence

The certainty of evidence for each primary outcome was assessed using the GRADE approach, considering risk of bias, inconsistency, indirectness, imprecision, and publication bias. Overall certainty was categorized as high, moderate, low, or very low. Given the predominance of retrospective designs, heterogeneity in AI methodologies, and limited external validation, the certainty of evidence for most diagnostic performance outcomes was rated as low to moderate.

RESULTS

Study Selection

The electronic database search identified a total of 312 records. After the removal of 84 duplicate records, 228 unique articles remained for title and abstract screening. During this stage, 198 studies were excluded because they were not related to artificial intelligence, did not involve fetal ultrasound imaging, focused on postnatal diagnosis, or did not address congenital heart defects.

Full-text assessment was performed for the remaining 30 articles. Of these, 25 studies were excluded for the following reasons: absence of major congenital heart defect outcomes (n = 9), focus exclusively on minor CHDs (n = 6), lack of diagnostic performance data (n = 5), postnatal imaging–based analysis (n = 3), or non–artificial intelligence computational approaches (n = 2).

Ultimately, five peer-reviewed studies met all predefined inclusion criteria and were included in the qualitative synthesis.

The study selection process is summarized in the PRISMA flow diagram. (Figure 1)

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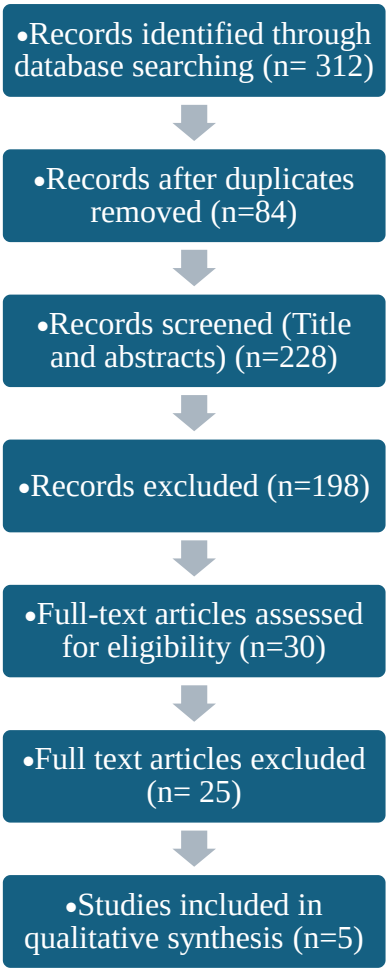

Figure 1: PRISMA Flowchart

Characteristics of Included Studies

Five peer-reviewed studies published between 2021 and 2026 were included in the final analysis. All studies evaluated deep learning–based artificial intelligence models applied to fetal ultrasound or fetal echocardiography for the detection of major congenital heart defects or ultrasound findings concerning severe cardiac pathology.

The included studies were conducted across diverse clinical settings, including tertiary referral centers, community-based screening programs, and multicenter cohorts. Imaging modalities ranged from routine second-trimester anomaly scans and cine ultrasound clips to targeted fetal echocardiography and first-trimester cardiac imaging. Reference standards included an expert fetal cardiologist interpretation, pediatric cardiologist adjudication panels, and postnatal diagnostic confirmation.

The characteristics of the included studies are summarized in Table 1.

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Review Article

Table 1: Characteristics of Included Studies

Author (Year)	Study Setting	Study Design	Imaging Modality	AI Approach	Target Condition	Reference Standard	Key Findings
Arnaout et al. (2021)[35]	Multicenter, tertiary	Retrospective	Fetal ultrasound and fetal echocardiography	Ensemble convolutional neural network	Complex/major CHDs	Expert fetal cardiologists	Expert-level diagnostic performance (AUC ≈ 0.99)
Tang et al. (2023)[36]	Multicenter	Retrospective-prospective	Fetal echocardiography (aortic arch view)	Two-stage transfer learning CNN	Duct-dependent CHDs	Expert fetal cardiologists	AUC ranging from 0.85 to 0.94 across datasets
Athalye et al. (2024)[37]	Community screening	Retrospective	Routine second-trimester anomaly scans	Convolutional neural network	Severe CHDs	Postnatal diagnosis and expert review	Sensitivity of 91%, outperforming routine screening
Lei et al. (2025)[38]	Multiregional	Retrospective	First-trimester fetal cardiac ultrasound	Interpretable deep learning model	CHD-suspicious cardiac patterns	Multicenter expert validation	High diagnostic accuracy with model explainability
Zelop et al. (2026)[39]	Multicenter (11 centers)	Retrospective	Second-trimester ultrasound cine clips	CNN-based finding detection model	Ultrasound findings concerning major CHDs	Pediatric cardiologist adjudication panel	Sensitivity of 96.8% for severe CHDs

Diagnostic Performance of Artificial Intelligence Models

Across all included studies, artificial intelligence models demonstrated consistently high diagnostic performance for detecting major congenital heart defects or ultrasound findings concerning severe cardiac pathology. Reported sensitivities ranged from approximately 84% to 96.8%, while specificity generally exceeded 90% where reported.

Studies utilizing multicenter datasets and external validation cohorts demonstrated improved robustness and generalizability across different ultrasound systems, imaging protocols, and operator expertise. In several studies, AI performance was comparable to that of expert fetal cardiologists and superior to routine clinical screening performed by non-specialist operators, particularly in community-based and low-risk screening settings.

Notably, models designed to detect suspicious morphologic ultrasound findings rather than attempting definitive lesion-specific diagnosis showed particularly high sensitivity for identifying fetuses requiring specialist referral.

Risk of Bias Assessment

The risk of bias of the included studies was assessed using a QUADAS-2 framework[34] adapted for artificial intelligence–based diagnostic accuracy studies. Overall, the risk of bias was judged to be low to moderate across studies. The most common sources of potential bias were related to retrospective study design, enriched prevalence of congenital heart defects, and use of stored ultrasound images rather than real-time scanning. In contrast, all studies employed appropriate and clearly defined reference standards, and the application of the AI index test was generally well described.

The results of the risk of bias assessment are summarized in Table 2.

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Review Article

Table 2: Risk of Bias Assessment

Study	Patient Selection	Index Test	Reference Standard	Flow and Timing	Overall Risk of Bias
Arnaout et al. (2021)[35]	Low	Low	Low	Moderate	Low-Moderate
Tang et al. (2023)[36]	Low	Low	Low	Low	Low
Athalye et al. (2024)[37]	Moderate	Low	Low	Moderate	Moderate
Lei et al. (2025)[38]	Moderate	Low	Low	Moderate	Moderate
Zelop et al. (2026)[39]	Low	Low	Low	Low	Low

DISCUSSION

The present study demonstrated that artificial intelligence (AI)-based approaches applied to fetal ultrasound imaging consistently improved the detection of ultrasound findings concerning major congenital heart defects (CHDs), addressing long-recognized limitations of conventional prenatal cardiac screening. Across the included studies, AI systems showed high diagnostic performance, reproducibility, and clinical applicability, particularly in screening environments where operator dependence, time constraints, and variability in expertise continue to limit prenatal detection rates.

Historical population-based evidence has repeatedly shown that prenatal detection of major CHDs remains suboptimal despite routine anomaly scanning. Early regional data from Sharland et al. (1992) [40] revealed that a substantial proportion of significant CHDs were missed during routine screening, even within organized healthcare systems. Long-term outcome data from Wren et al. (2008) [41] further demonstrated that, over 20 years, improvements in ultrasound technology alone did not lead to consistently high prenatal detection of life-threatening neonatal cardiovascular malformations. Similarly, large multicenter registry analyses by Garne et al. (2001) [42] and population-based screening studies by Tegnander et al. (2006)[43] highlighted wide inter-institutional variability in detection rates, underscoring the dominant influence of operator expertise, systematic view acquisition, and structured cardiac assessment.

Traditional strategies to improve prenatal CHD detection have focused on optimizing sonographic techniques and standardizing cardiac views. Carvalho et al. (2002) [44] demonstrated that inclusion of outflow tract assessment significantly improved detection of major CHDs compared with reliance on the four-chamber view alone. Yagel et al. (2001)[45] proposed the five short-axis view approach to facilitate comprehensive fetal cardiac evaluation, while subsequent reviews by Bravo-Valenzuela et al. (2018)[46] and Sun (2021)[47] emphasized that many critical CHDs manifest as subtle morphologic abnormalities easily overlooked in routine screening. These principles have been consolidated in international guidelines, including the updated ISUOG Practice Guidelines for fetal cardiac screening (Carvalho et al., 2023) [48], which emphasize systematic multiview assessment but also acknowledge persistent challenges related to operator dependence and real-world workflow limitations.

The included AI studies directly address these long-standing barriers by translating established screening principles into automated, reproducible systems. Arnaout et al. (2021) [35] demonstrated that an ensemble deep learning model integrating multiple standard fetal cardiac views achieved expert-level performance in detecting complex CHDs. This multiview strategy closely parallels the comprehensive evaluation advocated by Yagel et al. (2001) [45] and ISUOG guidelines, while reducing reliance on individual operator expertise. Athalye et al. (2024) [37] extended this approach into community-based screening, demonstrating that AI substantially improved the detection of severe CHDs in low-risk populations. This finding directly correlates with deficiencies identified in population-level studies by Sharland et al. (1992) [40], Garne et al. (2001)[42], and Tegnander et al. (2006)[43], suggesting that AI may be particularly valuable where specialist fetal cardiology expertise is limited.

While Arnaout et al. (2021) [35] and Athalye et al. (2024) [37] focused on broad screening for major CHDs, Tang et al. (2023) [36] adopted a more targeted strategy by evaluating AI performance for duct-dependent lesions using fetal echocardiography. Duct-dependent CHDs are associated with high neonatal morbidity and mortality when undiagnosed prenatally, as highlighted in outcome studies

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Review Article

by Wren et al. (2008) [41]. The lesion-focused, view-specific approach employed by Tang et al. (2023) [36] complements broader screening models and aligns with earlier physiologic and Doppler-based fetal cardiac assessment principles described by Rizzo et al. (1992) [49], demonstrating that AI frameworks can be tailored for both generalized screening and high-risk lesion detection.

A key conceptual shift observed across the included studies is the move away from lesion-specific diagnosis toward identification of ultrasound findings concerning major CHDs, a strategy that reflects real-world screening objectives. Zelop et al. (2026) [39] exemplified this approach by analyzing second-trimester cine clips to identify morphologic features associated with major CHDs, achieving very high sensitivity. This finding-based referral paradigm aligns with screening philosophies emphasized by Carvalho et al. (2002) [44] and Bravo-Valenzuela et al. (2018) [46], where the primary aim of routine ultrasound is timely recognition of abnormal findings that warrant specialist referral rather than definitive diagnosis at the screening stage.

The potential of AI to extend effective screening into earlier gestational windows is highlighted by Lei et al. (2025) [38], who demonstrated the feasibility of interpretable deep learning models for first-trimester fetal cardiac assessment. Early fetal echocardiography has historically been constrained by small cardiac size and technical limitations, with variable detection rates reported by Huggon et al. (2002) [50] and Smrcek et al. (2006) [51]. By providing both diagnostic outputs and visual explanations, interpretable AI models address concerns related to transparency and clinical trust, consistent with broader discussions on explainable AI in healthcare by Litjens et al. (2017) [52] and Esteva et al. (2019) [53].

Supporting technical literature further contextualizes the performance of the included AI systems. Baumgartner et al. (2017) [54] and Chen et al. (2015) [55] demonstrated that deep learning algorithms can reliably identify standard fetal cardiac planes in freehand ultrasound acquisition, addressing a critical upstream limitation in screening workflows. More recent studies by Yan et al. (2024) [56], Komatsu et al. (2021) [57], and Dozen et al. (2020)[58] showed that AI can accurately segment cardiac structures and vessels from fetal ultrasound videos, while Taksoee-Vester et al. (2024)[59] and He (2023)[60] highlighted the role of AI-based quality assessment in improving the consistency and reliability of fetal echocardiography. These upstream capabilities likely contribute to the high diagnostic performance observed in the included screening models and support the concept of integrated, quality-aware AI pipelines.

Despite these promising findings, several methodological limitations warrant consideration. Most included studies were retrospective and relied on stored images or cine clips, introducing potential spectrum bias and limiting assessment of real-time clinical performance. These issues are well recognized in diagnostic accuracy research, as outlined by Whiting et al. (2011) [34]. The emergence of AI-specific quality assessment and reporting frameworks, including QUADAS-AI (Sounderajah et al., 2021) [61] and TRIPOD guidelines (Collins et al., 2015) [62], underscores the importance of standardized methodology, transparent reporting, and prospective validation in future research.

In summary, the present study demonstrated that AI-based analysis of fetal ultrasound represents a coherent and clinically relevant evolution of prenatal cardiac screening. The included studies collectively illustrate complementary strategies, ranging from multiview screening and community-based implementation to targeted detection of duct-dependent lesions and early gestational assessment. When interpreted alongside extensive supporting evidence from fetal cardiology and AI methodology literature, these findings support the integration of AI as a screening adjunct aimed at improving detection consistency, facilitating timely referral, and potentially enhancing perinatal outcomes. Future prospective multicenter studies evaluating real-world workflow integration and clinical impact will be essential to define the optimal role of AI in routine prenatal care.

CONCLUSION

This systematic review demonstrated that artificial intelligence–based analysis of fetal ultrasound had substantial potential to improve the detection of ultrasound findings concerning major congenital heart defects by addressing key limitations of conventional prenatal screening, including operator dependence, variability in expertise, and inconsistent assessment of standard cardiac views. Across the included studies, AI models consistently showed high diagnostic performance in diverse clinical settings and functioned effectively as screening adjuncts aimed at identifying abnormal cardiac patterns warranting specialist referral rather than providing definitive diagnoses. Evidence from supporting fetal cardiology and imaging studies reinforced the clinical rationale for AI-assisted screening, particularly in community-based and low-risk

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Review Article

populations where prenatal detection rates had historically remained suboptimal. Despite these encouraging findings, the predominance of retrospective study designs and methodological heterogeneity highlighted the need for prospective, multicenter validation studies focusing on real-world workflow integration, clinical impact, and perinatal outcomes. Overall, artificial intelligence emerged as a promising adjunctive tool with the potential to enhance the consistency, equity, and effectiveness of prenatal screening for major congenital heart defects when implemented within robust clinical and methodological frameworks.

Limitations

This review has several limitations. First, heterogeneity in study design, AI algorithms, ultrasound protocols, and outcome reporting limited quantitative synthesis. Second, most included studies were retrospective and single-center, increasing the risk of selection bias and limiting generalizability. Third, variability in reference standards and lack of standardized reporting frameworks for AI diagnostic studies constrained comparability. Finally, publication bias cannot be excluded, as studies reporting favorable AI performance may be preferentially published.

Implications for Practice and Policy

The findings suggest that AI-assisted fetal ultrasound may enhance early detection of major congenital heart defects, particularly in settings with limited access to expert fetal cardiology services. However, current evidence does not yet support routine clinical implementation. Policymakers and regulatory bodies should emphasize the need for robust external validation, transparent reporting, and ethical oversight before widespread adoption. Integration into clinical workflows should be guided by evidence demonstrating improved diagnostic accuracy, clinical outcomes, and cost-effectiveness.

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LIST OF ABBREVIATIONS

AI – Artificial Intelligence
AUC – Area Under the Curve
CHD – Congenital Heart Defect
CNN – Convolutional Neural Network
GRADE – Grading of Recommendations Assessment, Development and Evaluation
PRISMA – Preferred Reporting Items for Systematic Reviews and Meta-Analyses

AUTHOR BIOGRAPHY

Dr. Suvarna Palanivelu contributed to the study conception, clinical interpretation of findings, and manuscript drafting.
Dr. Karthik Shunmugavelu conducted the literature search, data extraction, methodological appraisal, critical revision of the manuscript, and approved the final version.
Both authors reviewed and approved the final manuscript.

AUTHOR CONTRIBUTIONS

Conceptualization and study design: M.K., S.R.
Systematic literature search and study selection: M.K., P.V.
Data extraction and qualitative synthesis: M.K., S.R.
Risk of bias assessment and certainty of evidence evaluation: S.R.
Manuscript drafting: M.K.
Critical revision for important intellectual content: M.K., S.R., P.V.
All authors contributed substantially to the work, reviewed the final manuscript, and approved it for submission.

REGISTRATION AND PROTOCOL

This systematic review was not prospectively registered with PROSPERO.

SUPPORT

This study received no specific financial or non-financial support. The funders had no role in study design, data collection, analysis, interpretation, or manuscript preparation.

COMPETING INTERESTS

The authors declare no competing interests.

AVAILABILITY OF DATA, CODE, and OTHER MATERIALS

Extracted data supporting the findings of this study are available from the corresponding author upon reasonable request. No analytic code was generated, as this review did not involve primary statistical modeling. Data collection

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Review Article

forms and extracted datasets are not publicly archived but can be shared for academic purposes.

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Review Article

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