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Review Article

Comparison of the Accuracy of Prenatal and Postnatal Screening for the Detection of Congenital Heart Disease: A Systematic Review

Fadiyah Ulfah Khalid[1]

[1]*Department of Cardiology and Vascular, Faculty of Medicine, Hasanuddin University, Makassar, Indonesia*

Corresponding Author:
Name: Fadiyah Ulfah Khalid
Email: fadiyah.ts@gmail.com

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ABSTRACT

Introduction: *Congenital heart disease (CHD) is one of the most common congenital anomalies and remains a leading cause of neonatal morbidity and mortality worldwide. Early detection through prenatal and postnatal screening plays a crucial role in improving clinical outcomes; however, the diagnostic accuracy of these screening approaches varies across studies.* ***Method:*** *This systematic review was conducted and reported in accordance with the PRISMA 2020 guidelines. A comprehensive literature search was performed in PubMed, Embase, Web of Science, and the Cochrane Library up to December 20, 2025. Title, abstract, and full-text screening were independently conducted by two reviewers, with disagreements resolved through discussion. Eligible studies were English-language articles published between 2015 and 2025 that reported the diagnostic accuracy of prenatal screening (ultrasonography or fetal echocardiography) and/or postnatal screening (pulse oximetry or clinical examination) for CHD detection. Risk of bias and methodological quality were assessed using the Quality Assessment of Diagnostic Accuracy Studies-2 (QUADAS-2) tool. Due to substantial heterogeneity among studies, data were synthesized narratively.*

Conclusions: *Prenatal and postnatal screening methods exhibit complementary strengths in the detection of CHD. The implementation of an integrated two-stage screening strategy (prenatal–postnatal) is recommended as an optimal approach to enhance early detection and reduce neonatal morbidity and mortality associated with CHD, particularly in low- and middle-income countries.*

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1. INTRODUCTION

Congenital heart disease (CHD) refers to structural abnormalities of the heart that arise during embryonic development and encompass a wide spectrum of cardiac anatomical defects. Globally, CHD is recognized as one of the most common congenital anomalies and represents a leading cause of morbidity and mortality among newborns. The incidence of CHD is estimated to range from 8 to 10 cases per 1,000 live births; however, this figure varies across geographical regions and is influenced by differences in health surveillance systems, diagnostic methods, and reporting practices employed in individual studies.[1–3] Beyond its contribution to neonatal mortality, CHD imposes a substantial burden through long-term disability, the need for ongoing medical care, and significant economic costs, particularly in low- and middle-income countries.[1,3,4]

The high global prevalence and disease burden of CHD underscore the critical importance of early detection as an integral component of efforts to reduce neonatal mortality. Early identification of CHD enables optimal clinical planning, including appropriate selection of delivery location, preparedness of neonatal intensive care facilities, and timely implementation of surgical or medical interventions. Numerous studies have demonstrated that delayed diagnosis of CHD is associated with an increased risk of severe complications and neonatal death.[1,4] Therefore, the effectiveness of screening strategies plays a pivotal role in mitigating the adverse outcomes associated with CHD.

In general, CHD screening is conducted at two main stages: the prenatal period and the postnatal period. Prenatal screening primarily involves obstetric ultrasonography and fetal echocardiography, aiming to detect structural cardiac abnormalities as early as the second trimester of pregnancy. In contrast, postnatal screening relies on physical examination of the newborn and the use of pulse oximetry to identify hypoxemia, which may indicate the presence of critical CHD.[4,5] Although both approaches have been widely recommended, their diagnostic effectiveness and accuracy have shown considerable variability across studies.

Several studies report that prenatal screening offers advantages in detecting severe CHD before birth, thereby facilitating more comprehensive perinatal management. However, the accuracy of prenatal screening is highly dependent on technical factors, operator expertise, equipment quality, and gestational age at the time of examination.[1,3,5] Conversely, postnatal screening particularly the combination of physical examination and pulse oximetry has been shown to enhance the detection of

CHD cases that were missed during prenatal screening, although it remains limited in identifying certain non-cyanotic cardiac defects.[2] This variability in diagnostic performance highlights existing gaps in evidence regarding the most effective screening methods for early CHD detection.

Given these considerations, a systematic review comparing the diagnostic accuracy of prenatal and postnatal CHD screening, particularly in terms of sensitivity and specificity, is warranted. The findings of such a review are expected to provide a robust scientific foundation for the development of more effective and context-specific screening strategies, especially in developing countries such as Indonesia, where resource constraints and disparities in access to maternal and neonatal healthcare services remain significant challenges.

2. METHODS

This study is a systematic review conducted and reported in accordance with the PRISMA 2020 (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) guidelines.

a. Literature Search Strategy

A comprehensive literature search was performed across several international electronic databases, including PubMed, Embase, Web of Science, and the Cochrane Library. The search was conducted up to the latest available date, 20 December 2025. The search strategy employed a combination of relevant keywords and Boolean operators, adapted to the specific requirements of each database. The general search string used was as follows:

(“congenital heart disease” OR “congenital cardiac anomaly” OR “CHD”)
AND
(“prenatal screening” OR “antenatal screening” OR “fetal echocardiography” OR “prenatal ultrasound”)
AND
(“postnatal screening” OR “pulse oximetry” OR “clinical examination”)
AND
(“diagnostic accuracy” OR “sensitivity” OR “specificity” OR “predictive value”)

This search strategy was designed to ensure transparency and reproducibility, as well as to identify all relevant studies evaluating the diagnostic accuracy of prenatal and postnatal screening for congenital heart disease (CHD).

b. Inclusion and Exclusion Criteria

The inclusion criteria for this systematic review were as follows:

1. Studies published within the last 10 years (2015–2025);
2. Articles published in the English language;
3. Full-text articles available for review;
4. Observational or diagnostic study designs, including cross-sectional, cohort, or retrospective studies;
5. Studies reporting prenatal screening (fetal echocardiography or prenatal ultrasound) and/or postnatal screening (pulse oximetry or clinical examination) for the detection of CHD;

6. Studies providing data on diagnostic accuracy parameters, such as sensitivity, specificity, positive predictive value (PPV), negative predictive value (NPV), or overall diagnostic accuracy.

Grey literature including unpublished reports, preprints, theses, dissertations, and conference abstracts was excluded from this review. These sources were excluded because they generally lack rigorous peer-review processes, which may compromise methodological validity and the reliability of reported diagnostic accuracy outcomes.

c. Study Selection Process

Study selection was conducted in a stepwise manner. The first stage involved screening titles and abstracts to identify potentially relevant studies. Studies that met the initial eligibility criteria were subsequently assessed through full-text review to confirm their inclusion. The study selection process is presented using a PRISMA 2020 flow diagram.

d. Data Extraction

Data were systematically extracted from each included study using a predefined extraction framework. The following information was collected:

1. Author(s) and year of publication;
2. Country and study design;
3. Type of screening method (prenatal or postnatal);
4. Timing of screening;
5. Sample size;
6. Number of CHD cases detected;
7. Diagnostic accuracy measures, including sensitivity, specificity, PPV, and NPV.

e. Risk of Bias Assessment

The methodological quality and risk of bias of the included studies were assessed using the Quality Assessment of Diagnostic Accuracy Studies-2 (QUADAS-2) tool. This instrument evaluates four key domains:

1. Patient selection;
2. Index test;
3. Reference standard;
4. Flow and timing.

Each domain was rated for risk of bias (low, high, or unclear), as well as concerns regarding applicability. This assessment was conducted to ensure that the reported sensitivity and specificity estimates were derived from studies with adequate methodological rigor.

f. Data Analysis

Data analysis was primarily conducted using a qualitative (narrative) synthesis, comparing the diagnostic accuracy of prenatal and postnatal screening modalities. Where sufficient homogeneity across studies was observed, a meta-analysis was planned to pool sensitivity and specificity estimates using a random-effects model. In cases of substantial heterogeneity between studies, findings were summarized descriptively through narrative synthesis.

3. RESULTS

The database search identified a total of 2,969 articles. After the removal of 569 duplicate records, 2,400 articles were screened based on titles and abstracts, of which 2,320 were excluded due to irrelevance. A total of 80 full-text articles were subsequently assessed for eligibility, and 65 articles were excluded because they did not report adequate diagnostic accuracy data, employed inappropriate study designs, involved non-relevant populations, or were published outside the predefined inclusion period. Ultimately, 10 studies met the inclusion criteria and were included in this systematic review.

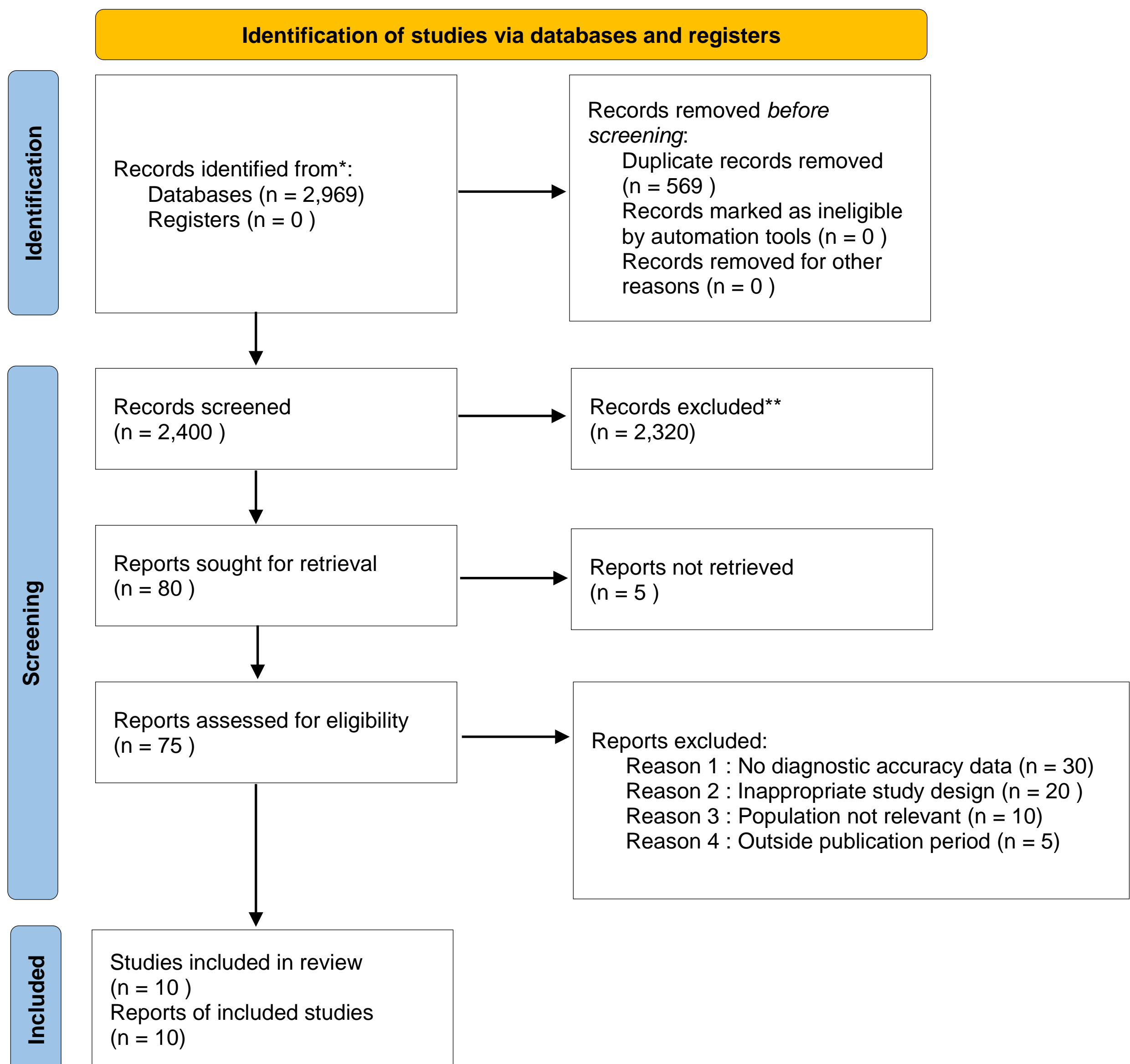

Figure 1. Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) Flow Diagram.

The articles were presented in a tabular format containing information such as the author's name, year of publication, title, publisher, country of origin, and key

findings. Data extracted from the journals that met the inclusion criteria were compiled into **Table 1**.

Table 1. Research Findings from Studies Included in the Systematic Review

No	Author (Year)	Title	Country	Key Findings
1	Mamalis et.al (2023)	*Comparison of the Results of Prenatal and Postnatal Echocardiography and Postnatal Cardiac MRI in Children with a Congenital Heart Defect*	Germany	Retrospective–prospective study. Fetal echocardiography showed sensitivity of 90–100% and specificity of 97–100%. Postnatal MRI clarified complex vascular anatomy. High prenatal accuracy for major CHD, except for DORV.
2	Plana et,al (2018)	*Pulse Oximetry Screening for Critical Congenital Heart Defects (Cochrane Review)*	United Kingdom	Meta-analysis of 21 studies (457,202 newborns). Sensitivity 76.3% (95% CI 69–82), specificity 99.9% (95% CI 99.7–99.9). False positive 0.14%. POS is highly specific and effective for early CCHD detection.
3	Slitine et.al (2020)	*Pulse Oximetry and Congenital Heart Disease Screening: Results of the First Pilot Study in Morocco*	Morocc	Detected 5 CCHD cases; failure rate 0.18%; one false negative; supports routine CCHD screening
4	Murni et al. (2022)	*Feasibility of Screening for Critical Congenital Heart Disease Using Pulse Oximetry in Indonesia*	Indonesian	Cross-sectional study in Yogyakarta. Sensitivity 75.9%, specificity 98.8%. POS feasible for national implementation with healthcare training and simple equipment.
5	Jullien S. (2021)	*Newborn Pulse Oximetry Screening for Critical Congenital Heart Defects*	Spain	Policy and evidence review. Concluded POS consistently accurate with sensitivity 76%, specificity 99.9%. Recommends integration with antenatal and clinical screening.
6	Singh Y., Chen	*Impact of Pulse Oximetry Screening to Detect*	United Kingdom	Five-year prospective study. Sensitivity 85.7%, specificity

No	Author (Year)	Title	Country	Key Findings
	S.E. (2022)	*Congenital Heart Defects: 5 Years' Experience in a UK Regional Neonatal Unit*		99.3%. Two-thirds of major CHDs were missed prenatally, but all CCHDs were detected. POS reduced neonatal mortality.
7	Majani et al. (2025)	*Pulse Oximetry Screening for Critical Congenital Heart Disease in Tanzanian Newborns: Diagnostic Accuracy, Sensitivity, and Specificity in a Low-Resource Healthcare Setting*	Tanzania	Prospective cohort of 10,630 newborns. Sensitivity 50%, specificity 99.5%. Accuracy depended on screening time (optimal at 48–72 hours). POS feasible in low-resource settings.
8	Jawin et.al (2015)	*Beyond Critical Congenital Heart Disease: Newborn Screening Using Pulse Oximetry for Neonatal Sepsis and Respiratory Diseases in a Middle-Income Country*	Malaysia	Prospective observational study. Sensitivity 100%, specificity 99.7% for CCHD; also detected respiratory disease and sepsis. POS is cost-effective for dual neonatal screening.
9	Ding J., et al. (2023)	*Sensitivity and Specificity of Second-Trimester Anatomy Ultrasound for Detection of Fetal Congenital Heart Disease*	China	Prenatal diagnostic study. Sensitivity 86.5%, specificity 99.4%. Accuracy improved with operator experience and high-resolution equipment. Important for prenatal screening comparison.
10	Janjua D., Singh J., Agrawal A. (2022)	*Pulse Oximetry as a Screening Test for Congenital Heart Disease in Newborns*	India	Multicenter observational study. Sensitivity 80.6%, specificity 99.1%. Combining POS with physical examination improved CCHD detection to 100%. Recommended for medium-resource hospitals.

4. DISCUSSION

The findings of this systematic review demonstrate that prenatal and postnatal screening play complementary roles in the detection of congenital heart disease (CHD), particularly critical congenital heart disease (CCHD). Numerically, prenatal screening using fetal echocardiography and second-trimester anatomical

ultrasonography shows higher sensitivity, ranging from 85% to 100%, with specificity between 97% and 100% (Mamalis et al., 2023; Ding et al., 2023).[6,7] In contrast, postnatal screening using pulse oximetry screening (POS) exhibits more variable sensitivity, ranging from 50% to 86%, while maintaining consistently high specificity of 98–99.9% across most studies (Plana et al., 2018; Singh & Chen, 2022; Janjua et al., 2022).[8–10] This comparison highlights that prenatal screening is superior for early detection of major structural cardiac anomalies, whereas postnatal screening excels in minimizing false-positive results and ensuring that critical cardiac defects are not missed after birth.

Despite these advantages, substantial heterogeneity in sensitivity and specificity estimates was observed across studies. One of the primary sources of heterogeneity is operator dependency, particularly in prenatal screening methods. Studies by Mamalis et al. (2023) and Ding et al. (2023) demonstrated that the sensitivity of fetal echocardiography increased significantly in tertiary referral centers with experienced operators and access to high-resolution imaging equipment. Nevertheless, complex cardiac anomalies such as double outlet right ventricle (DORV) were still frequently missed, underscoring the inherent limitations of prenatal screening in identifying certain anatomical configurations. These findings reinforce the notion that prenatal diagnostic accuracy is highly contingent upon examiner expertise and facility quality.[6,7]

Additional heterogeneity arises from population characteristics and health system contexts. Studies conducted in high-income countries, such as the United Kingdom and Sweden, reported higher POS sensitivity (80–86%) compared with studies from low- and middle-income countries (LMICs), including Tanzania and Indonesia, where sensitivity ranged from approximately 50% to 76% (Murni et al., 2022; Majani et al., 2025).[11,12] These discrepancies are likely attributable to differences in CHD prevalence, access to confirmatory diagnostic tools such as echocardiography, and the readiness of neonatal referral systems.

The timing of screening is another critical factor influencing the diagnostic performance of POS. Majani et al. (2025) explicitly demonstrated that POS sensitivity increased significantly when screening was performed at 48–72 hours after birth compared with screening conducted before 24 hours of life. Early screening is associated with an increased risk of false-negative results due to the persistence of physiological postnatal shunts. This finding underscores the importance of standardized screening timing, particularly in resource-limited healthcare settings.[12]

Although prenatal screening offers higher sensitivity, postnatal screening provides notable advantages, including simplicity, non-invasiveness, low cost, and feasibility for widespread implementation. Evidence from Plana et al. (2018) indicates that integrating prenatal and postnatal screening more than doubles the detection rate of CCHD compared with using either method alone. This is further supported by Mamalis et al. (2023), who reported very high agreement between prenatal and postnatal findings (Cohen’s kappa >0.9). Collectively, these findings suggest that an optimal screening strategy should be complementary rather than substitutive, combining both prenatal and postnatal approaches.[6]

Furthermore, evidence from LMIC settings strengthens the case for broad implementation of POS. A pilot study by Slitine et al. (2020) in Morocco demonstrated

that POS detected CCHD with a very low failure rate (0.18%) and only one false-negative case, supporting the feasibility of routine POS implementation even in resource-constrained environments.[13] Additionally, Jullien (2021), in a WHO Europe policy review, emphasized the global consistency of POS accuracy and recommended its integration with antenatal screening and routine neonatal clinical examination to maximize CCHD detection.[14] These findings are consistent with the study by Jawin et al. (2015) in Malaysia, which reported high sensitivity and specificity of POS for CCHD and highlighted additional benefits, including early detection of neonatal respiratory disease and sepsis. Collectively, these studies confirm that POS is a feasible, cost-effective screening intervention with broad clinical impact, particularly in health systems with limited resources.[15]

This systematic review has several limitations that should be acknowledged. First, substantial heterogeneity existed across studies in terms of study design, screening timing, outcome definitions, and diagnostic confirmation methods, limiting the ability to perform direct quantitative comparisons. Second, variability in study designs including observational studies, cohort studies, diagnostic accuracy studies, and meta-analyses may have influenced the consistency of sensitivity and specificity estimates. Third, a meta analysis was not conducted due to methodological heterogeneity and inconsistent data reporting, resulting in a narrative synthesis of findings. Moreover, many studies did not report detailed risk of bias assessments, which may affect the interpretation of diagnostic accuracy estimates. These limitations highlight the need for future well designed, standardized prospective studies with uniform reporting to strengthen the evidence base.

4. CONCLUSIONS

This systematic review demonstrates that both prenatal screening using fetal echocardiography and postnatal screening using pulse oximetry exhibit high accuracy in detecting congenital heart disease (CHD), particularly critical congenital heart disease (CCHD). Prenatal screening provides higher sensitivity in identifying complex structural cardiac anomalies and plays a crucial role in delivery planning and preparedness for immediate neonatal intervention. In contrast, postnatal pulse oximetry screening shows very high specificity, is non-invasive, cost-effective, and easy to implement, especially in healthcare settings with limited resources.

The integration of these two modalities as a two-tier screening strategy yields the most optimal detection outcomes and has the potential to reduce delays in diagnosis, morbidity, and mortality associated with CHD. These findings have important implications for national health policies, particularly in low- and middle-income countries (LMICs), where limited access to fetal echocardiography may be compensated by the implementation of universal neonatal pulse oximetry screening. Therefore, the integration of CCHD screening into national maternal and child health programs should be strongly considered, accompanied by the strengthening of referral systems and the availability of confirmatory diagnostic services.

Further research is warranted to support evidence-based policy implementation, including cost-effectiveness analyses of integrated screening strategies, evaluations of training-based interventions for healthcare providers to enhance detection accuracy, and implementation science studies to assess the sustainability, acceptability, and

overall impact of CCHD screening programs across diverse healthcare system contexts. Such approaches are expected to accelerate the adoption of effective and equitable CHD screening strategies and ultimately improve neonatal health outcomes on a global scale.

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Conflict of Interest Statement:

The author declares that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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