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Sensitivity and Specificity of Pulse Oximetry for Congenital Heart Disease Screening in Newborn: A Meta-Analysis

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ABSTRACT

Background: Early detection for Congenital Heart Disease (CHD) using pulse oximetry is a routine procedure for newborn. Pulse Oximetry Screening (POS) has been shown to be effective in detecting CHD.
Aims: To evaluate the accuracy of POS through sensitivity and specificity in detecting CHD.
Methods: In this meta-analysis, we conducted a search on Pubmed, Scopus, and ScienceDirect for studies that were published up to June 20, 2024. We selected studies that assessed the sensitivity and specificity of POS for the screening of CHD in newborn babies in a hospital or home setting, regardless of gestational age at birth and excluded newborns with a previous diagnosis of CHD. We used RevMan5 software (QUADAS-2: Quality Assessment of Diagnostic Accuracy Studies-2) to assess the quality of the studies included and used Stata/SE 16.0 software to pool the sensitivity and specificity.
Results: The results showed that 17 studies, including 413,516 newborns, had pooled sensitivity and specificity of 0.69 (95% CI: 0.57-0.81) and 0.93 (95% CI: 0.85-1.00).
Conclusion: This study's results suggest that POS has moderate sensitivity and high specificity in screening CHD in newborns. It can be concluded that there is a prospective usefulness of POS as a valuable tool in clinical practice for screening and management of CHD, especially in settings where access to higher diagnostic technologies may be limited.

Keywords: *Congenital heart disease, Newborn, Pulse oximetry*
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1. Introduction

Congenital heart disease (CHD) is one of the most prevalent congenital diseases among newborns. It has a significant number of morbidity and mortality. Screening as an early detection of CHD is very important, as an early intervention can lead to better outcomes for affected infants. Pulse Oximetry Screening (POS) is a non-invasive, also cost-effective, screening tool that measures oxygen saturation and it has been selected as a method for CHD screening in newborns.

Recent studies have shown the importance of integrating POS with more advanced diagnostic tools to enhance diagnostic rates of CHD. For example, combining POS with physical examination and echocardiography can show a more comprehensive evaluation in newborns, and could potentially identify cases that might be missed when only performing POS as a single tool (Mannan et al., 2022). Furthermore, more studies showed, that it is important to consider many factors, such as gestational age and birth weight, as they may influence the interpretation of POS, particularly in preterm newborns or newborns in high altitudes, where adjusted thresholds are recommended (Nathawani et al., 2024). This multi-faceted approach not only supports the importance of early diagnosis but also highlights the need for standardized protocols for screening.

POS now emerges as a non-invasive and cost-effective tool for the early detection of CHD in newborns. This technique measures the oxygen saturation levels, diagnosing hypoxemia, which is a common indicator in cardiopulmonary disorders. The introduction of POS as a screening method has a potential approach in detecting CHD, especially in settings where advanced diagnostic technologies are not present (Minocha et al., 2018).

The integration of advanced diagnostic technologies into POS protocols could further enhance the CHD detection capabilities. For example, utilizing new-generation pulse oximetry that has better performance in detecting precise oxygen saturation, could enhance the accuracy of the screening (Anne de-Wahll Granellii, 2009). Another study revealed an incorporation of machine learning algorithms to summarize the patterns of oxygen saturation that may allow physicians to identify a newborn with possible CHD more effectively, thus allowing early intervention. Regarding some studies that showed many life-threatening conditions still undetected in initial physical examination, adopting such innovative tools could be helpful in reducing morbidity and mortality associated with undiagnosed CHD (Nathawani et al., 2024). This screening methodology evolution not only shows the importance of improving newborn care but also shows the necessity for elevating the knowledge among the healthcare professionals.

This meta-analysis aims to know the effectiveness of POS in detecting CHD in newborns from various studies. By knowing the sensitivity and specificity of pulse oximetry, we seek to identify a difference between current literature and offer recommendations for future study. The goal of this study is to contribute to the latest discussion on enhancing CHD screening protocols.

2. Methods

Search strategy

This systematic review and meta-analysis was conducted to know the sensitivity and specificity of pulse oximetry in screening for CHD in newborns. The review procedure followed the PRISMA guidelines to make sure the review is comprehensive and transparent. This study used electronic databases, including PubMed, ScienceDirect, and Scopus. The search strategy included keywords and medical subject headings (MeSH) terms related to "congenital heart disease," "pulse oximetry," and "newborn screening." The search was limited to articles published in English up to June 20th, 2024.

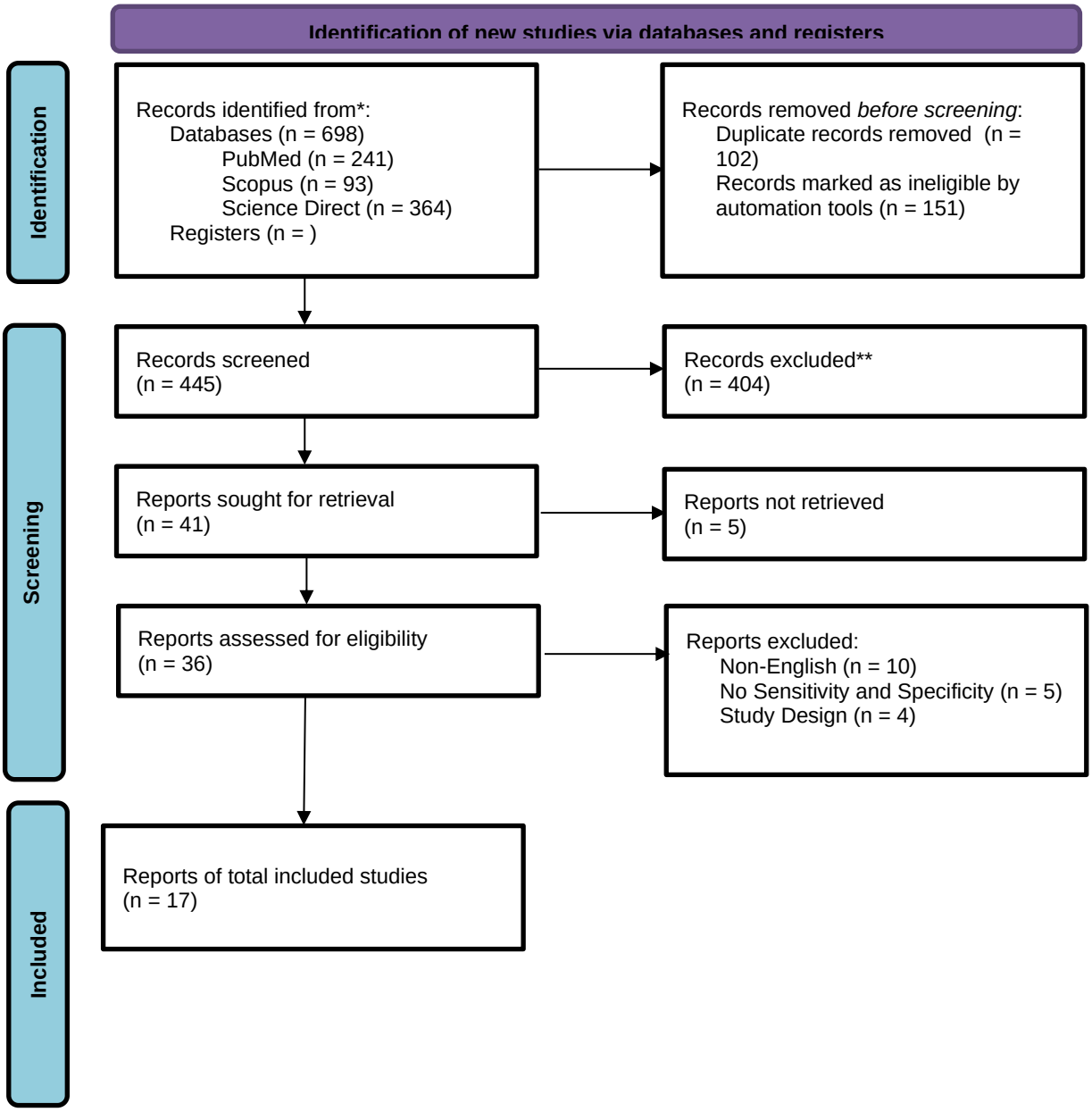

Figure 1. *The PRISMA study selection flowchart. This flowchart shows the number of records and studies at each stage of the study selection process.*

Criteria for inclusion and exclusion

Studies were included if:

1. Evaluated the effectiveness of pulse oximetry as a screening tool for CHD in newborns.
2. Reported on sensitivity and specificity
3. Involved a population of newborns within the first month of life.

Studies were excluded if:

1. Newborn with previous diagnosis of Congenital Heart Disease.

Search Result

We assessed the titles and abstracts of 698 records identified from electronic databases. Of these records, we identified 17 studies—published between up to June 2024—as eligible for inclusion (Fig. 1).

Quality Assessment

The methodological quality of the included studies was assessed using the QUADAS-2 tool, which evaluates the risk of bias and applicability of the studies. Each study was classified based on the domains of patient selection, index test, reference standard, and flow and timing.

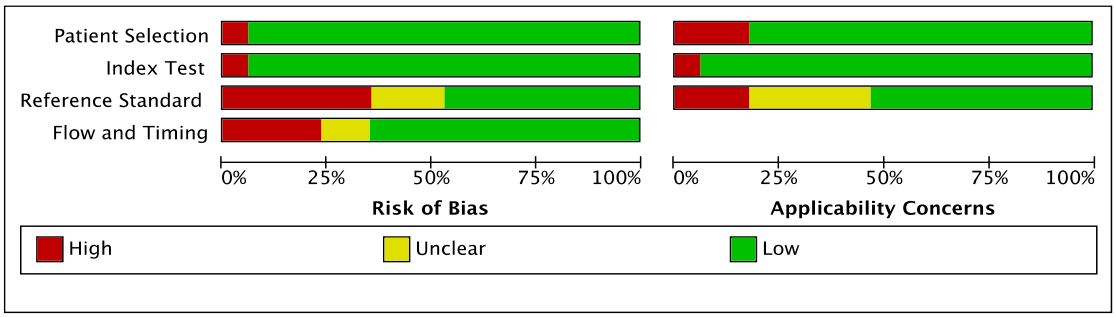

Figure 2 *Distribution of risk of bias and applicability concerns across studies*

For risk of bias, most studies demonstrated a low risk in the domain of Patient Selection. Only one study, Riede FT et al 2009, had a high risk of bias, while all other studies were rated as low risk. In the Index Test domain, most studies also showed a low risk of bias; however, there were exceptions (Figure 2). Miller et al 2016 and Singh et al 2022 were rated as having a high risk of bias, and Mathur NB et al 2015 were deemed unclear. The Reference Standard domain presented a more variable picture, with studies like Al-Mawazini et al 2017 and Schena et al 2017 marked as high risk, while others, such as Azhar AS et al 2018, had a low risk of bias. Some of the studies had an unclear risk in this domain. Regarding flow and timing, most studies were categorized low risk, but studies from Al-Mawazini et al 2017 and Miller et al 2016, were categorized high risk, indicating potential biases in the flow and timing domain (Figure 3).

In the applicability concerns, most studies have low concerns in the patient selection domain. Similarly, in the index test domain, most of the studies are categorized into low applicability concerns, except one study, Bradshaw et al 2012, which is categorized into unclear applicability concerns. In the reference standard domain, few studies, like Siefkes et al 2024, are categorized into unclear applicability concerns (Figure 3).

The quality assessment shows most studies are categorized into low risk of bias and low applicability concerns in patent selection domain and index test domain. However many studies are categorized into high risk and unclear risk of bias and applicability concerns in reference standard domain and flow and timing domain. These findings warn us to be more careful in extracting data from studies with a high risk of bias.

3. Results

This study examined the sensitivity and specificity of POS in different populations from 17 studies included. Using the random-effects REML model, the pooled sensitivity is 0.69 (95% CI: 0.57–0.81). The sensitivity across individual studies ranged from as low as 0.04 (95% CI: 0.02–0.06) in Huang et al. to as high as 0.99 (95% CI: 0.98–1.00) in Zhao et al. Heterogeneity was substantial, with an I^2 of 98.71%, showing high variability among the included studies. The τ^2 value was 0.05, and the Q statistic was significant (Q[16] = 6972.11, p = 0.00), further confirming heterogeneity. Despite the variation across studies, the overall pooled sensitivity suggests moderate diagnostic accuracy (Figure 4). Studies contributing the most weight to the overall estimate included Zhao et al. (7.10%), Ewer et al. (7.00%), and Huang et al. (7.09%). In contrast, studies such as Miller KK et al. (4.00%) and Bradshaw et al. (4.80%) contributed the least weight. The variation in weights reflects the sample size or precision of individual study estimates.

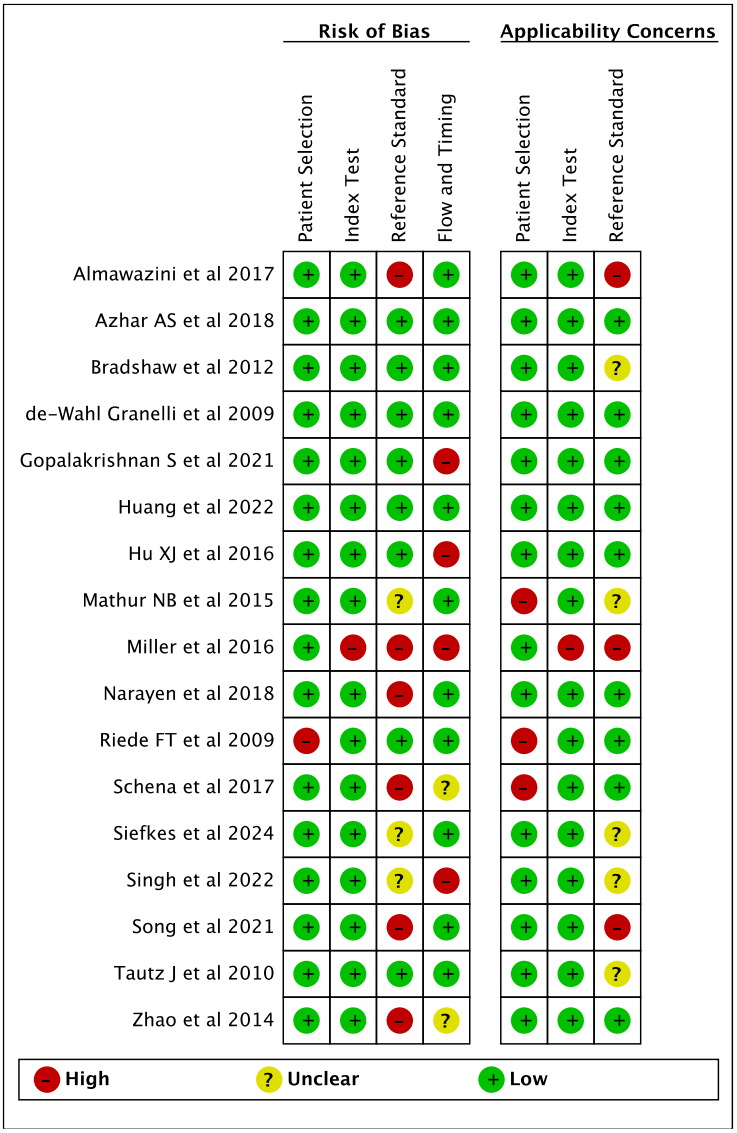

Figure 3 *Detailed risk of bias and applicability concerns assessment for individual studies*

The pooled sensitivity, calculated using a random-effects REML model, was 0.69 (95% CI: 0.57–0.81), reflecting a moderate level of diagnostic accuracy. Sensitivity estimates across individual studies exhibited considerable variation, ranging from 0.04 (95% CI: 0.02–0.06) in Huang et al. to 0.99 (95% CI: 0.98–1.00) in Zhao et al. Significant heterogeneity was observed among the studies, as evidenced by an I^2 value of 98.71%, which highlights substantial variability between studies. The τ^2 value was 0.05, and the Q statistic (Q[16] = 6972.11, p = 0.00) confirmed the presence of this heterogeneity. Despite this variability, the majority of studies demonstrated moderate-to-high sensitivity estimates. Notable contributions came from studies such as Singh et al. (0.82, 95% CI: 0.76–0.88), Hu XJ et al. (0.94, 95% CI: 0.80–1.08), and Mathur NB et al. (0.89, 95% CI: 0.81–0.97), which carried weights between 6.49% and 6.98%. These studies played a significant role in the overall pooled sensitivity. In contrast, studies with lower sensitivity, such as Huang et al. (0.04, 95% CI: 0.02–0.06) and Narayen et al. (0.50, 95% CI: 0.19–0.81), had smaller contributions to the overall analysis due to their limited diagnostic performance. Despite the high degree of heterogeneity, the pooled sensitivity highlights the moderate diagnostic capability of the included studies (Figure 4).

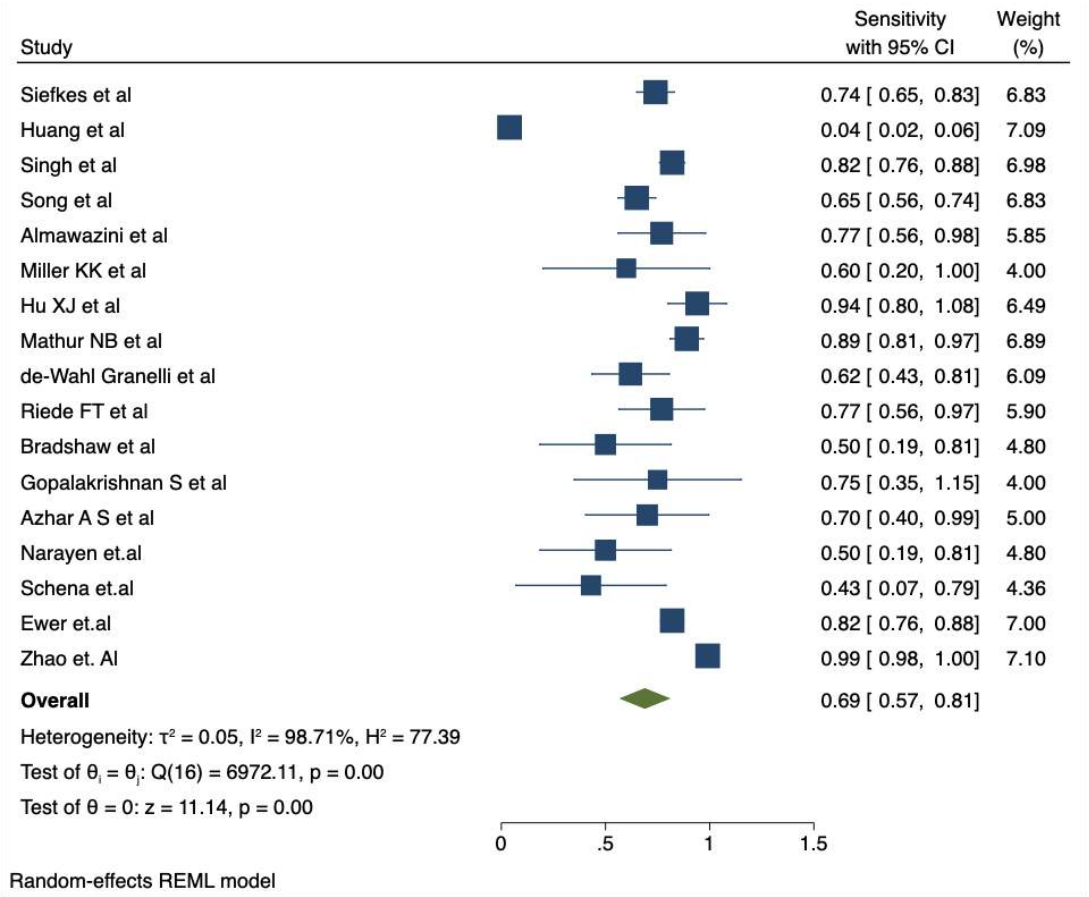

Figure 4 *Forest plot displaying the sensitivity estimates (with 95% confidence intervals) for 17 studies assessing the diagnostic accuracy of pulse oximetry*

The pooled specificity, estimated using a random-effects REML model, was 0.93 (95% CI: 0.85–1.00). Specificity estimates across individual studies were generally high, ranging from 0.40 (95% CI: 0.38–0.42) in Hu XJ et al. to 0.99 (95% CI: 0.98–1.00) in several studies, including Huang et al., Singh et al., Miller KK et al., and Zhao et al. The heterogeneity of the studies was significant, with an I^2 of 99.89%, indicating a high level of variability between studies. The τ^2 value was 0.03, and the Q statistic (Q[16] = 3526.73, p = 0.00) confirmed the presence of heterogeneity. Despite this, the overall specificity suggests excellent diagnostic performance across the studies (Figure 3). Most of the studies, including Singh et al., Riede FT et al., and Schena et al., demonstrated high specificities (close to or at 0.99), contributing equally to the overall estimate with weights around 5.90%. The studies by Hu XJ et al. (5.88%) and Mathur NB et al. (5.74%) exhibited lower specificity estimates, impacting their relative weights in the overall analysis (Figure 5).

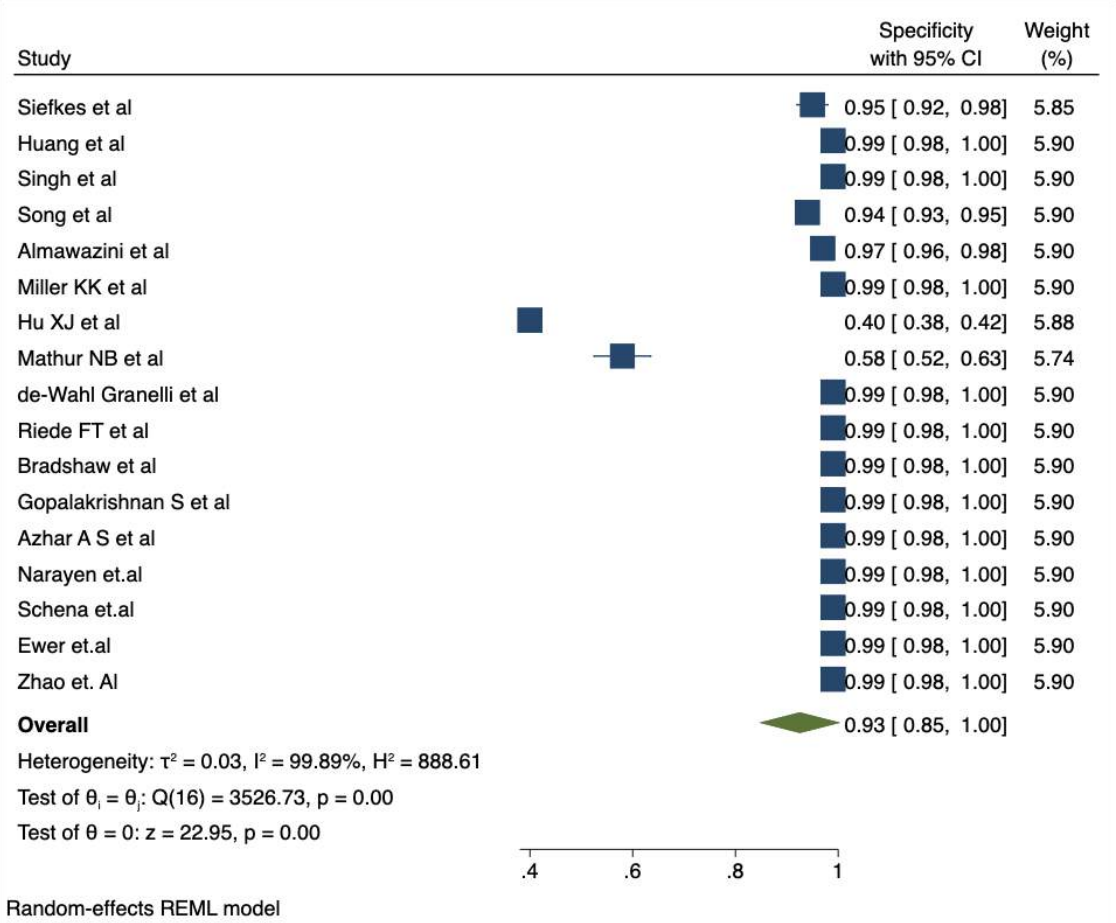

Figure 5 *Forest plot displaying the specificity estimates (with 95% confidence intervals) for 17 studies assessing the diagnostic accuracy of pulse oximetry*

4. Discussion

This systematic review and meta-analysis provide an in-depth look into the diagnostic performance of pulse oximetry for congenital heart disease (CHD) screening in newborns. The pooled sensitivity of 0.69 (95% CI: 0.57–0.81) indicates that pulse oximetry identifies approximately 69% of true CHD cases, highlighting its moderate sensitivity. POS can detect many CHD cases but may miss a significant number. The pooled specificity is 0.93 (95% CI: 0.85–1.00), which shows that POS is good in ruling out CHD in healthy newborns, so the false positives and unnecessary follow-up procedures could be reduced. This combination of moderate sensitivity and high specificity emphasizes pulse oximetry's role as an effective initial screening method rather than a standalone diagnostic tool for CHD in newborns.

The high heterogeneity observed in sensitivity (I^2 = 98.71%) and specificity (I^2 = 99.89%) reveals considerable variability in diagnostic performance across studies. Different study populations, screening protocols, the timing of POS, and the type of pulse oximetry used, could lead to a high heterogeneity score. For example, the studies held in advanced diagnostic technologies settings with standardized protocols may show higher sensitivity and specificity, while studies held in limited settings may face challenges affecting accuracy. Timing of the POS conducted also could interfere with the oxygen saturation reading, conducting POS too early, could show hypoxemia that is not related to CHD. According to these factors, in future studies, a clarification of the screening protocols is needed (Gopalakrishnan et al., 2021; Riede et al., 2010; Schena et al., 2013).

In this analysis, larger studies with specific estimates—such as those by Zhao et al. and Ewer et al.—carried more weight in the pooled results, strengthening the reliability of the overall estimates for sensitivity and specificity. In contrast, smaller studies, or those with less precise estimates, like Hu XJ et al., had minimal influence on the pooled results due to lower weights assigned in the meta-analysis. Recognizing the impact of study size and precision on pooled estimates provides insight into how certain studies contribute to overall findings, emphasizing the value of larger, well-designed studies in informing clinical practice.

The diagnostic value of pulse oximetry in newborn CHD screening appears promising given its high specificity, as it can reliably exclude CHD in infants without the condition. However, with moderate sensitivity, it may not detect all cases, suggesting that pulse oximetry alone may not suffice for a definitive diagnosis. Instead, it is better suited as an initial screening tool that, when combined with additional diagnostic tests such as echocardiography, could improve the accuracy and comprehensiveness of newborn CHD screening. This combined approach may be particularly useful in settings where early CHD detection is crucial for timely intervention.

To enhance pulse oximetry's utility in CHD screening, further research should aim to standardize protocols and evaluate its effectiveness across diverse settings and populations. Additionally, exploring combinations with other screening tools, such as echocardiography, could help to optimize sensitivity and overall diagnostic accuracy. Future studies could also investigate adjustments in timing, cutoff values, and device selection to better accommodate different clinical contexts. By addressing these areas, further research could help to refine pulse oximetry screening and maximize its impact on early CHD detection and neonatal health outcomes.

5. Conclusion

In conclusion, although pulse oximetry has a high specificity, the sensitivity is still moderate. Considering the results, POS could be a prospective valuable screening tool. It is non-invasive and cost-effective, suitable to be used in a setting where access to advanced diagnostic technology is limited.

Conflict of Interest

There is no conflict of interest. Nothing to disclosure

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