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Diagnostic Accuracy of Trans-Vaginal Ultrasound for Deep Infiltrative Endometriosis: A systematic review

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Abstract

Introduction: Deep infiltrative endometriosis (DIE) is a severe form of endometriosis, affecting approximately 20% of women with the disease. It often goes undiagnosed due to its complex presentation and overlap with other pelvic conditions. Early and accurate detection is key to guiding treatment and reducing the disease burden. Transvaginal ultrasound (TVUS) is a first-line imaging method used in clinical settings due to its accessibility and low cost. Despite this, reported accuracy in identifying DIE remains inconsistent.

Objective: This review aims to assess the pooled sensitivity and specificity of Trans-Vaginal Ultrasound (TVUS) for diagnosing Deep Infiltrative Endometriosis (DIE).

Methods: A systematic search of PubMed, Web of Science, and Scopus was conducted to identify relevant studies published between 2012 and 2024. Eligible studies included those that evaluated the diagnostic accuracy of TVUS for DIE, using surgical and histopathological findings as reference standards. Methodological quality was assessed via the QUADAS-2 tool.

Results: Sensitivity of TVUS ranged from 26.7% to 98%, while specificity varied from 46% to 100%. The highest sensitivity noted to be 98%, and the lowest to be 26.7%. Several studies, reported specificity of 100%. Among multicenter prospective studies, reported accuracy values were of 94% and 86.5%, respectively.

Conclusion: This systematic review reports the diagnostic utility of TVUS in detecting DIE, with many studies reporting high levels of sensitivity, specificity, and overall accuracy. The variation observed across individual studies emphasizes the impact of methodological differences, imaging techniques, and examiner

proficiency on diagnostic outcomes. These findings highlight the need for continued efforts to standardize imaging practices and enhance training to ensure consistent and reliable use of ultrasound in clinical settings.

Keywords: Diagnostic techniques and procedures, ultrasonography, endometriosis.

Introduction

Deep infiltrative endometriosis (DIE) is a severe form of endometriosis, affecting approximately 20% of women with the disease. It often goes undiagnosed due to its complex presentation and overlap with other pelvic conditions. Early and accurate detection is key to guiding treatment and reducing the disease burden. Transvaginal ultrasound (TVUS) is a first-line imaging method used in clinical settings due to its accessibility and low cost. Despite this, reported accuracy in identifying DIE remains inconsistent. In more advanced cases, it may also involve distant anatomical sites, including the bowel, bladder, and, on rare occasions, extra-pelvic regions such as the lungs [1].

Deep infiltrating endometriosis (DIE) often presents with symptoms that can be both severe and disruptive to daily life. Women commonly report chronic pelvic pain, especially around menstruation, along with deep dyspareunia (pain during intercourse), painful bowel movements, and urinary discomfort, depending on the organs involved. In some cases, infertility may be the first sign that leads to further investigation. The intensity of symptoms doesn't always match the extent of the disease, which can make diagnosis challenging. Since DIE can affect multiple pelvic structures like the bowel, bladder, and uterosacral ligaments, the clinical presentation is often complex and requires a high index of suspicion from clinicians [1,2,3].

Laparoscopic visualization, followed by histologi-

cal confirmation, has traditionally been regarded as the gold standard for diagnosing endometriosis. However, its invasive nature, high cost, and potential complications contribute to diagnostic delays, often spanning several years from the onset of symptoms. These limitations underscore the critical need for reliable, non-invasive diagnostic alternatives [3].

Transvaginal ultrasound (TVUS) has emerged as a promising first-line imaging modality in the evaluation of suspected endometriosis. Compared to other diagnostic tools, TVUS is widely accessible, cost-effective, well-tolerated by patients, and free of ionizing radiation. Advancements in ultrasound technology and growing operator proficiency have markedly enhanced the ability to detect and characterize endometriotic lesions, particularly deep infiltrating endometriosis (DIE)—a severe subtype associated with deep tissue invasion and significant morbidity [4,5,6].

This systematic review aims to synthesize the current evidence on the diagnostic performance of TVUS in identifying endometriosis. By evaluating the sensitivity, specificity, and accuracy reported across studies, the review seeks to provide clinicians with a comprehensive understanding of the diagnostic utility of TVUS in routine gynecological practice and inform future research directions.

Methodology

This systematic review was conducted in accordance with the Preferred Reporting Items for Sys-

tematic Reviews and Meta-Analyses (PRISMA) guidelines [7]. A comprehensive literature search was performed across three major databases PubMed, Web of Science, and Scopus to identify studies published between 2012 and 2024 that evaluated the diagnostic performance of transvaginal ultrasound (TVUS) in detecting deep infiltrative endometriosis. In the absence of a pooled meta-analysis, we presented the diagnostic performance of ultrasound for DIE using a scatter plot of sensitivity versus specificity across 28 studies based on Cochrane Handbook for Systematic Reviews of Diagnostic Test Accuracy which scatter plots to show sensitivity/specificity [47,48].

Eligibility Criteria

Studies were eligible for inclusion if they:

- Assessed the diagnostic accuracy of TVUS for DIE
- Used surgical visualization and/or histopathological confirmation as reference standards
- Reported data on sensitivity, specificity, or diagnostic accuracy;
- Were published in English between 2012 and 2024.

Articles were excluded if they involved insufficient sample size, lacked diagnostic outcome measures, or employed non-comparable methodologies.

Study Selection

An initial total of 500 records was retrieved from the database search. After removing 100 duplicates, 400 studies were screened based on title and ab-

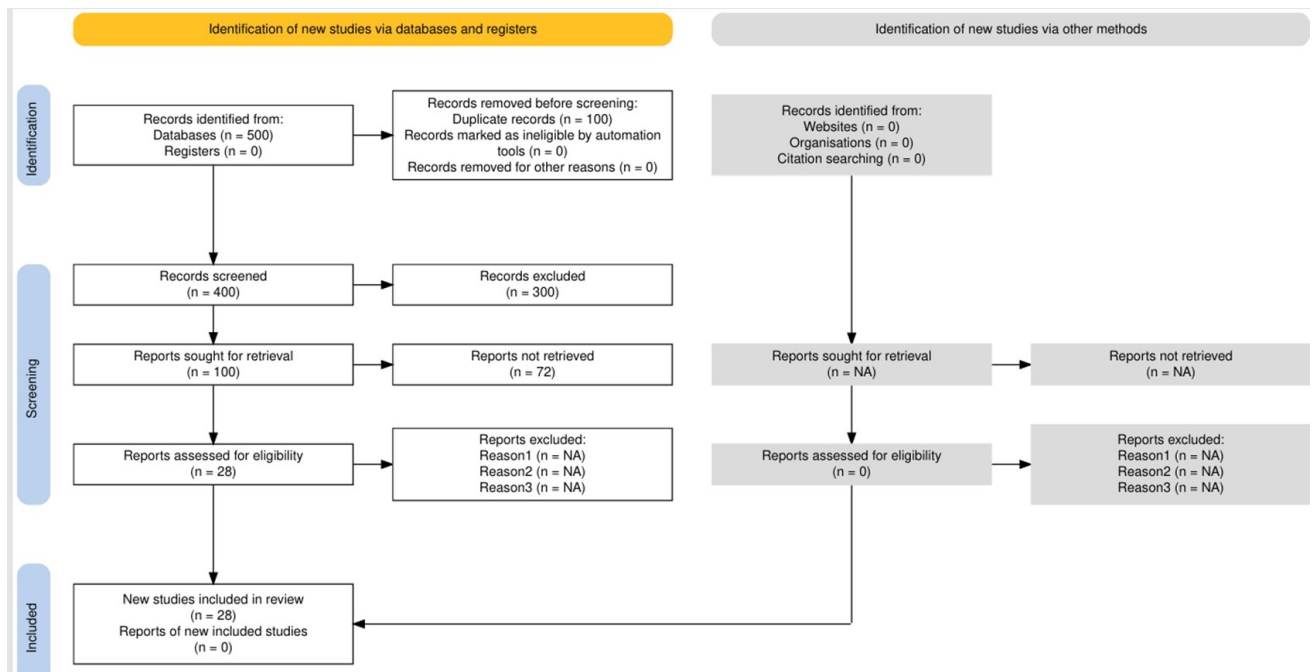


Figure 1. Flowchart of studies included in systematic review.

Table 1. Summary of pooled results of sensitivity, specificity and diagnostic accuracy of ultrasound for detection of DIE.

Sr. no.	Author (Year)	Study design	Reference standard	Population size	Sensitivity	Specificity	Accuracy	Lesion location
1	Padmehr et al., 2023	retrospective cross-sectional study		170	86.76% & 70.86%	86.76% & 70.86%	100%	For left ovarian & right ovarian endometriosis
2	Savelli et al., 2012	prospective design	Surgery and histopathology	381	77% & 45%	95% & 98%	88% & 81%	Overall values for expert operators & first-level operators
3	Leonardi et al., 2022	international, multicenter prospective	Surgery and histopathology	273	89.8% & 88.4%	75.9% & 78.8%	85.9% & 86.1%	Overall pooled values based on histology & surgical visualization
4	Abrão et al., 2007	cross-sectional design	MRI	104	98% & 95%	100% & 98%	99% & 97%	With respect to the rectosigmoid and retro cervical sites
5	Fernandez et al., 2024	retrospective observational case series	Surgery and histopathology	100	NA	NA	92%	Cul-de-sac, rectovaginal septum, uterosacral ligament, cervix, ovaries
6	Chen et al., 2019	retrospective longitudinal study	Surgery and histopathology	29	26.7% & 42.9%	85.7% & 87.5%	55.2%	With respect to identification of rectal infiltration & rectovaginal endometriosis
7	Baušić et al., 2023	retrospective longitudinal study	Laparoscopy and histopathology	256	93.78%	57.14%		Ovarian endometriomas
8	Vimercati et al., 2012	Prospective study	Laparoscopy and histopathology	90	81.1%	94.2%	89.2%	Overall
9	Bartlett et al., 2020	Retrospective observational study	Laparoscopy and histopathology	83	22% by US 61% by MRI	Not provided	22%	Overall
10	Freger et al., 2023	Prospective study, secondary analysis		54	82.6%, 75.0%, 100%	100%, 100%, 100%	92.6%, 94.4%, 100%	For the left USL, right USL, and torus uterine (TU)
11	Chen et al., 2024	Prospective study	Laparoscopy	42	92.3%	93.8%	92.9%	For DIE nodules on USLs with and without POD fluid
12	Xiang et al., 2022	Systematic review and meta-analysis	Laparoscopic surgery, histopathology	1,707	98%	100%	97%	Overall pooled values
13	Ros et al., 2021	Prospective study	Laparoscopic surgery, histopathology	172	96.6% & 83.9%	82.1% & 89.4%	89.5% & 86.6%	For USLs & TU

continued

Table 1. Continued

14	Gonçalves et al., 2021	Prospective study	Laparoscopy	120	85.7%	99.1%	92.4%	For retrocervical, ovarian, and bladder endometriosis
15	Leonardi, 2022	Prospective international pilot study	Surgery, histopathology	273	88.4%	78.8%	86.1%	Overall values based on surgery
16	Montanari, 2022	Prospective multicenter study	Surgery, histopathology	745	95%	93%	94%	For rectovaginal septum and vagina
17	Kamkarfar et al., 2022	Comparative evaluation	Surgery, histopathology	80	93%	65%	79%	Endometriosis based on laparoscopy
18	Moradi et al., 2022	Comparative evaluation	Laparoscopy	69	88.6%	60%	74.3%	DIE nodule
19	Sadighi et al., 2023	Cross-sectional study	Laparoscopy, histopathology	110	58.3%	98.7%	89.5%	For DIE and POD obliteration in the anterior pelvic compartment
20	Guerriero et al., 2016	Systematic review and meta-analysis	Surgery	2639	91%	97%	94%	Rectosigmoid
21	Guerriero et al., 2015	Systematic review and meta-analysis	Surgery	1583	53%	93%	73%	Overall pooled values
22	Keckstein et al., 2022	Prospective, multicenter study	Surgery	745	95% (#Enzian A)	99% (#Enzian FI)	99% (#Enzian FB)	Enzian compartments
23	Hudelist et al., 2011	Systematic review and meta-analysis	Laparoscopy, histopathology	1106	91%	98%	94.5%	Bowel endometriosis
24	Gerges et al., 2021	Systematic review and meta-analysis	Laparoscopy, histopathology	3374	89%	97%	93%	Pooled value for rectal/rectosigmoid DE
25	Phua et al., 2016	Observational cohort study	Laparoscopy	214	82%	97%	89.5%	POD obliteration
26	Deslandes et al., 2020	Systematic review	Laparoscopy, histopathology	35 studies	78.5% to 85.3%	46.1% to 92.5%	75.7% to 97%	Overall pooled values
27	Zhao et al., 2022	Meta-analysis	Surgery	23 studies	72%	99%	85.5%	Overall pooled values
28	Garzon et al., 2024	Prospective observational design	Laparoscopic surgery	476	90.74%	98.58%	97.69%	Parametria endometriosis

stract. Of these, 100 full-text articles were assessed for eligibility. Seventy two studies were excluded based on predefined criteria, such as lack of relevant diagnostic outcomes or inappropriate design, resulting in 28 studies being included in the final review.

Results

Study Selection and Characteristics: Out of approximately 500 records retrieved from the database search, 100 duplicates were removed. The remaining 400 articles underwent screening based on their titles and abstracts. After this initial screening, 100 studies were selected for full-text review. Of these, 72 articles were excluded due to reasons such as unsuitable study design, limited sample size, or incomplete data reporting. As a result, 28 studies met the inclusion criteria and were incorporated into the final review.

These selected studies represented a wide spectrum of research methodologies, including prospective and retrospective cohorts, cross-sectional investigations, observational studies, and several meta-analyses. The sample sizes across studies varied considerably, ranging from as few as 29 to as many as 3,374 participants. Where participant age was mentioned, the average age fell between 31.5 and 40 years. The PRISMA flow chart is as shown in Figure 1 [46].

The studies included in this review demonstrated a wide range of diagnostic performance for ultrasound in identifying endometriosis. Sensitivity values ranged from as low as 26.7% to as high as 98%, while specificity ranged between 46% and 100%. The lowest sensitivity (26.7%) was found in the study by Chen *et al.* (2019), whereas Xiang *et al.* (2022) reported the highest sensitivity at 98%. Specificity reached near-perfect levels 100% in several investigations [18,31].

Higher overall accuracy rates were frequently ob-

served in meta-analyses. For instance, different studies reported an accuracy of 93%, 94%, and 94.5% respectively. Among the prospective multicenter trials, a study demonstrated a high diagnostic accuracy of 94%, another study reported slightly lower accuracy of 86.5% respectively.

Table 1 shows summary pooled results of sensitivity, specificity and diagnostic accuracy of ultrasound for detection of endometriosis. Table shows that this systematic review included 28 studies evaluating the diagnostic performance of ultrasound in detecting deep infiltrating endometriosis. The studies span various designs, including prospective, retrospective, and meta-analyses, with sample sizes ranging from small cohorts to large-scale reviews involving thousands of patients. Sensitivity values varied widely across studies from as low as 22% to as high as 98% while specificity generally remained high, often exceeding 90%, particularly in meta-analyses and larger prospective designs. Accuracy rates also showed a strong trend toward reliability, with many studies reporting figures above 85%. Notably, the strongest diagnostic performance was observed in meta-analyses and large prospective studies.

Table 2 is the summary of the risk of bias assessment of the 28 included studies showed mixed results. Most prospective multicenter studies and meta-analyses had a low risk of bias, which makes their findings more reliable. On the other hand, several retrospective and smaller observational studies showed moderate to high risks of selection, performance, or detection bias. These issues mean that some studies have overestimated accuracy by including easier-to-diagnose cases, while others have underestimated it because of operator differences or lack of blinding.

The results of this review showed wide variation in diagnostic accuracy, which reflects heterogeneity among the included studies. This variability was

Table 2. QUADAS-2 Risk of Bias Assessment of the Included Studies

Sr. no	Author (Year)	Study Design	Sample Size	Selection Bias Risk	Performance Bias Risk	Detection Bias Risk
1	Padmehar et al., 2023	Retrospective cross-sectional	170	Moderate	Low	Low
2	Savelli et al., 2012	Prospective	381	Low	Moderate	Low
3	Leonardi et al., 2022	Multicenter prospective	273	Low	Low	Low
4	Abrão et al., 2007	Cross-sectional	104	High	Moderate	High
5	Fernandez et al., 2024	Retrospective observational	100	Moderate	High	Moderate
6	Chen et al., 2019	Longitudinal	29	High	High	High
7	Baušić et al., 2023	Retrospective longitudinal	256	Moderate	Moderate	Low
8	Vimercati et al., 2012	Prospective	90	Low	Low	Low
9	Bartlett et al., 2020	Retrospective observational	83	High	Moderate	High
10	Freger et al., 2023	Prospective secondary analysis	54	Low	Low	Low
11	Chen et al., 2024	Prospective	42	Low	Low	Low
12	Xiang et al., 2022	Systematic review/meta-analysis	1707	Low	Low	Low
13	Ros et al., 2021	Prospective	172	Low	Moderate	Low
14	Gonçalves et al., 2021	Prospective	120	Low	Low	Low
15	Wertz, 2022	International pilot study	-	High	High	Moderate
16	Montanari, 2022	Prospective multicenter	745	Low	Low	Low
17	Kamkarfar et al., 2022	Comparative evaluation	80	Moderate	Moderate	Moderate
18	Moradi et al., 2022	Comparative evaluation	69	Moderate	High	Moderate
19	Sadighi et al., 2023	Cross-sectional	110	High	Low	High
20	Guerrero et al., 2016	Systematic review/meta-analysis	2639	Low	Low	Low
21	Guerrero et al., 2015	Systematic review/meta-analysis	1583	Low	Moderate	Low
22	Keckstein et al., 2022	Prospective multicenter	745	Low	Low	Low
23	Hudelist et al., 2011	Systematic review/meta-analysis	1106	Low	Low	Low
24	Gerges et al., 2021	Systematic review/meta-analysis	3374	Low	Low	Low
25	Phua et al., 2016	Observational cohort	214	Moderate	Low	Low
26	Deslandes et al., 2020	Systematic review	35 studies	Moderate	High	Moderate
27	Zhao et al., 2022	Meta-analysis	23 studies	Low	Low	Low
28	Garzon et al., 2024	Prospective observational	476	Low	Low	Low

mainly due to differences in lesion location, study design, and the level of operator expertise. Although we did not perform a formal statistical test of heterogeneity, the broad range of sensitivity and specificity values in our review suggests that the findings are best understood within a random-effects framework. This highlights that clinical and methodological differences between studies strongly influence the reported diagnostic outcomes.

Figure 2 shows the scatter plot of sensitivity and specificity for transvaginal ultrasound (TVUS) in diagnosing deep infiltrating endometriosis (DIE) across included studies.

Each point represents an individual study, showing variability in diagnostic performance. Most stud-

ies demonstrated high sensitivity and specificity, while some reported lower sensitivity, reflecting differences in operator expertise, study design, or lesion location. This plot visually highlights the heterogeneity in diagnostic accuracy among the included studies.

Discussion

This systematic review reports the varying diagnostic accuracy of ultrasound in identifying endometriosis, with reported sensitivities ranging from 26.7% to 98% and specificities between 46% and 100%. Such a broad range in diagnostic performance likely reflects differences in study design, imaging

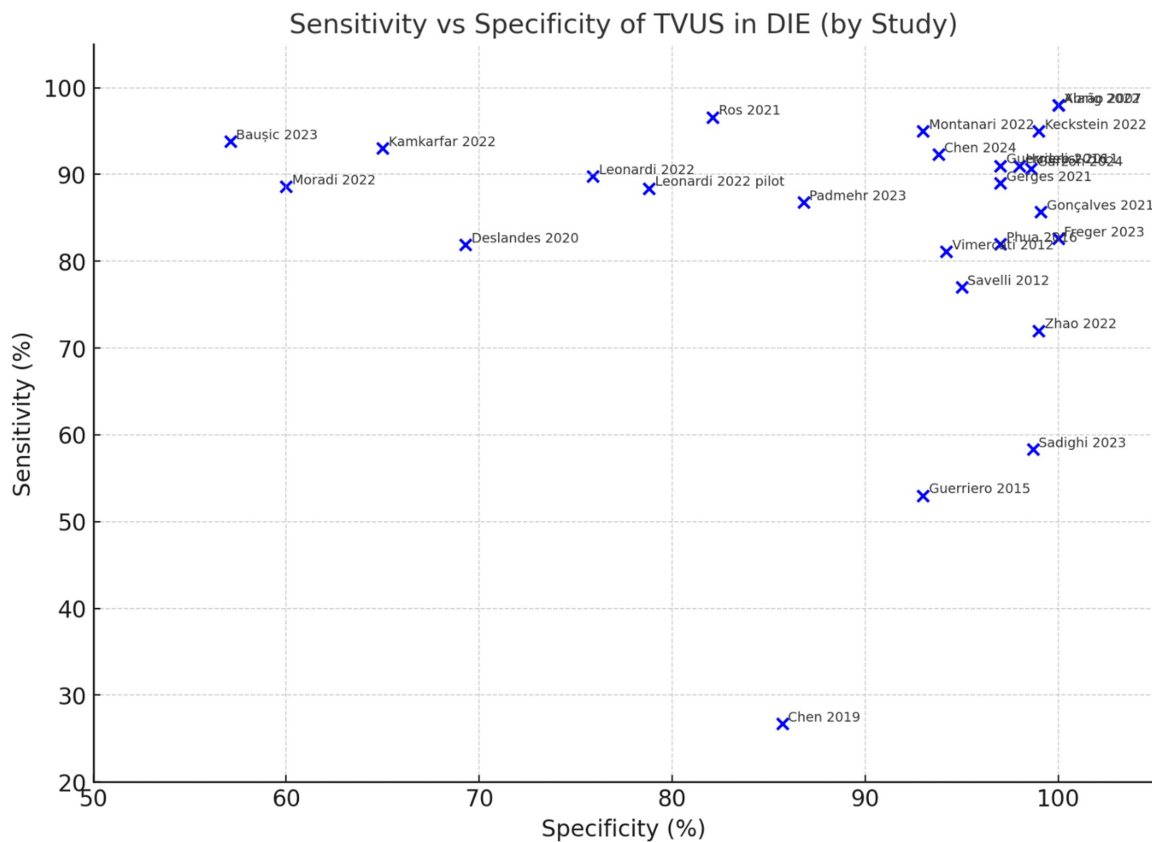


Figure 2. Scatter plot of sensitivity and specificity for transvaginal ultrasound (TVUS) in diagnosing deep infiltrating endometriosis (DIE) across included studies.

protocols, operator skill, and patient-related variables. Notably, meta-analyses which compile data across multiple studies tended to show higher sensitivity and specificity, suggesting that when ultrasound is performed using standardized approaches, it can serve as a reliable and non-invasive diagnostic method for endometriosis.

However, the variation in results across individual studies cannot be overlooked. For instance, a study reported a particularly low sensitivity (26.7%), pointing toward the influence of factors such as lesion location, limited sample size, non-standardized imaging techniques, or atypical disease presentations [14]. On the other hand, few studies demonstrated near-perfect specificity, highlighting ultrasound's potential to confidently rule out endometriosis when applied under optimal conditions [21,18].

A clear pattern also emerged when comparing prospective versus retrospective study designs. Prospective multicenter investigations showed more consistent and higher diagnostic accuracy [25,43]. In contrast, retrospective studies tended to exhibit greater variability, likely due to inconsistencies in data collection, reporting, and imaging criteria. Similarly, some systematic reviews and meta-analyses reported more robust diagnostic values, further emphasizing the strength of well-structured, large-scale research in establishing clinical benchmarks [29,34].

When interpreting the diagnostic performance of TVUS for DIE, it is important to consider its role relative to other imaging modalities, particularly MRI. Comparative studies have shown that TVUS offers diagnostic accuracy comparable to MRI for certain sites, such as rectosigmoid and ovarian endometriosis, but MRI may perform better in detecting lesions at more complex anatomical locations, including the uterosacral ligaments and bladder. In a study conducted by Bartlett et al in 2020 shows that 22 percent of endometriosis was detected by in 18 of 83

patients while MRI detected endometriosis in 61 percent of patients 51 of 83. In 51 percent of Patients DIE was not detected on US but positive on MRI 33 of 65 patients. MR identified additional sites or sequelae in the majority of patients [17].

Conclusion

In conclusion, this review shows that transvaginal ultrasound (TVUS) is a reliable and valuable tool for diagnosing deep infiltrating endometriosis, especially when performed by trained and experienced operators. Most well-designed studies reported good accuracy, making TVUS a strong first-line imaging option. However, some studies with higher risks of bias remind us that results should be interpreted carefully. MRI can provide additional information in complex cases, but because of its higher cost and limited availability, TVUS remains the most practical choice in routine practice.

Limitations

Forest plots could not be generated for all included studies due to unavailability of complete diagnostic 2x2 data and confidence intervals.

Conflict of Interest

None.

Grant support and financial disclosure

None.

Data availability

All data analyzed in this study were extracted from previously published articles, which are available in the public domain. No new data were generated.

Authors contribution

Fatima Batool: Concept, article writing database, literature search, PRISMA flow chart.

Javeria Khan: Literature search.

Dr Syed Muhammad Yousuf, Prof. Syed Amir Gillani, Dr. M Mudassar Mahmood ,Dr Khadija: Critical review, proof reading.

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