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Diagnostic Approach to Anorectal Malformations in Neonates

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Abstract

Anorectal malformation (ARM) is a congenital anomaly in neonates with an incidence of approximately 1 in 5,000 live births. The wide spectrum of abnormalities necessitates accurate classification and early diagnosis during the neonatal period to guide appropriate surgical management and predict long-term continence outcomes. One of the major diagnostic challenges lies in the limitations of conventional invasive techniques and the potential risks associated with radiation exposure. This study aims to identify effective diagnostic approaches for ARM in neonates. This research employs a literature review design with a narrative review approach. The data consist of secondary sources derived from 20 selected studies, including national and international journals as well as textbooks published between 2014 and 2025, accessed through databases such as ClinicalKey and Google Scholar. The analysis focuses on clinical, radiological, prenatal, and systemic factors relevant to establishing an accurate diagnosis. The findings indicate that diagnosis should begin with a thorough perineal physical examination to identify fistulas and signs of obstruction. A high-pressure distal colostogram remains the gold standard for delineating fistula anatomy prior to surgery. In addition, high-resolution transperineal ultrasound and magnetic resonance imaging have emerged as reliable non-invasive modalities for evaluating rectal position and the sphincter muscle complex. Anorectal manometry also plays a significant role in detecting subtle forms of ARM that may be overlooked during routine examinations. A systematic and multidisciplinary diagnostic approach that integrates clinical assessment, screening for associated anomalies, and multimodal imaging is essential to optimize surgical planning and improve patient outcomes.

Introduction

Anorectal malformations (ARM) are one of the most common congenital anomalies in neonates, with an incidence of approximately 1 in 5,000 live births (Stoll et al., 2007; Suseelan et al., 2026; Hasan et al., 2026). The spectrum of these anomalies is very broad, ranging from anal atresia without a fistula to complex forms involving the urogenital tract. Accurate diagnosis and classification during the neonatal period are critical as they determine the surgical management strategy and long-term prognosis regarding continence function. An accurate diagnostic approach allows for the identification of the fistula's anatomy and its relationship with surrounding structures, thereby minimizing the risk of postoperative complications (Jun et al., 2021; Wei et al., 2026; Shahid et al., 2026).

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To date, conventional methods such as the high-pressure distal colostogram have been considered the gold standard for assessing ARM types and detecting fistulas to the urogenital tract (Etskovitz et al., 2026; However, this examination is invasive, requires the use of contrast agents and radiation, and relies on the operator's experience to achieve optimal pressure without causing rupture or suboptimal results. These limitations create a need for a safer, non-invasive diagnostic method that can be repeated without the risk of radiation exposure in neonates (Oyania et al., 2024; Rabandiyarov et al., 2026; Liang et al., 2026).

Advances in imaging technology have enabled the use of high-resolution transperineal ultrasound, including three-dimensional (3D transperineal ultrasound) approaches, as a promising alternative for evaluating anorectal anatomy (Wang et al., 2026; Zakaria et al., 2025; Giannakodimos et al., 2025). This examination can visualize the location of the rectal tip, its relationship with the sphincter complex, and the presence of fistulas in real-time without radiation exposure. Additionally, this examination can be performed on neonates without requiring anesthesia or invasive procedures, making it an ideal choice in the early phase of assessment (Davidson et al., 2024; Olivieri et al., 2026; Hardcastle et all., 2025).

Although various studies have demonstrated the potential of transperineal ultrasound in assessing ARM, systematic research is still needed to determine its diagnostic accuracy compared to high-pressure distal colostomy as the gold standard (Xu et al., 2025). Research on diagnostic approaches for anorectal malformations in neonates is highly relevant for identifying the most effective, safe, and efficient methods in daily clinical practice. The integration of appropriate imaging results will support optimal surgical planning and improve patients' long-term functional outcomes (Oh et al., 2020).

Anatomical variations in anorectal malformations not only impact surgical correction techniques but also directly affect the development of defecation function and the patient's future quality of life. The complexity of the relationship between the rectal tip, sphincter muscles, and urogenital structures demands precise evaluation from the earliest stages of life. Errors in identifying the type of malformation can lead to suboptimal procedural choices, ultimately increasing the risk of incontinence, chronic constipation, or urological complications. Modern diagnostic focus is no longer solely on morphological classification but also on functional mapping oriented toward long-term outcomes (Pelizzo et al., 2023).

In this context, ultrasound-based approaches offer advantages in dynamically visualizing soft tissues, particularly in assessing the complex external sphincter and levator ani muscles. Real-time visualization capabilities allow clinicians to evaluate spatial relationships more comprehensively than static radiological techniques. 3D technology provides volumetric reconstructions that aid in understanding anatomical orientation more intuitively, particularly in cases with complex fistula configurations. This is crucial in determining whether a patient can undergo primary procedures such as posterior sagittal anorectoplasty (PSARP) without a colostomy stage (Gangopadhyay & Pandey, 2015).

Safety considerations are also a primary concern in the neonatal population, where radiation exposure has greater biological consequences than in other age groups. The use of ultrasound eliminates this risk while allowing for repeated evaluations to monitor development or plan staged interventions. This flexibility is particularly relevant in cases with unstable clinical conditions, where invasive procedures could potentially worsen the patient's overall condition. Additionally, the non-invasive approach enhances patient comfort and reduces the need for additional interventions such as sedation (Kuang et al., 2024).

From a clinical implementation perspective, the main challenges in adopting transperineal ultrasound are the need for standardization of techniques and improved operator competence. Variability in results can occur if the procedure is not performed according to consistent protocols, including patient positioning, probe pressure, and image interpretation. The

development of evidence-based clinical guidelines and structured training are key to ensuring that this method is widely reliable. Integration with other modalities, such as MRI in complex cases, can also enhance diagnostic accuracy in a multimodal approach (Paradiso et al., 2023).

Future research should focus on large-scale comparative studies evaluating the sensitivity, specificity, and predictive value of ultrasound compared to conventional methods. Cost-effectiveness analyses are also crucial for assessing the feasibility of implementation across various healthcare facilities, particularly in resource-limited settings in developing countries. This technology-driven approach has the potential to revolutionize ARM diagnostic algorithms by positioning safer and more efficient methods as the first line of evaluation, thereby supporting more precise and evidence-based clinical decision-making.

Methods

The study conducted is a Literature Review with a Narrative Review design. The data used in this study consists of secondary data, specifically post-experimental studies from various sources obtained via the internet, including research results from international journals, national journals, citations from the UMI Faculty of Medicine, Clinical Key, textbooks, and conference proceedings from 2014 to 2025. The narrative review approach offers flexibility in integrating various research findings with diverse methodological characteristics, both quantitative and qualitative. This model allows researchers to construct a comprehensive conceptual synthesis, particularly on topics that are still evolving and lack a standard consensus. By reviewing various literature sources, researchers can identify patterns, trends, and unresolved research gaps. This is particularly important in the context of anorectal malformations, where clinical variations and diagnostic approaches continue to evolve (Tuginem, 2023).

The selection of the publication timeframe also plays a role in ensuring that the review reflects current developments in the fields of imaging and pediatric surgery. More recent literature generally includes technological innovations, updates to clinical protocols, and research findings with better methodological quality. The process of selecting credible sources, such as reputable journals and verified scientific databases, contributes to the validity of the review's findings. Additionally, a critical evaluation of each source is necessary to assess the strength of evidence, potential biases, and relevance to the study's objectives.

Analysis in a narrative review is not merely descriptive but also interpretive, emphasizing the interconnections among findings. Researchers need to group study results based on specific themes, such as diagnostic accuracy, procedural safety, or clinical implications, thereby producing a systematic line of argumentation. This approach enables the formation of a conceptual framework that can serve as the basis for clinical practice recommendations and future research directions. A robust analytical structure enhances the scientific contribution of the review and clarifies the research's position within the broader scientific landscape.

Result and Discussion

The process of identifying and selecting articles in this study was conducted systematically. Based on the search results, a total of 124 articles were identified using the keyword "diagnostic approaches for anorectal malformations in neonates." All retrieved articles were initially included in the search record and then subjected to screening based on their relevance to the topic, publication period, accessibility of the full text, and suitability to the focus of the review. The researchers first screened the titles and abstracts to determine whether each article discussed diagnostic approaches, clinical evaluation, imaging modalities, associated anomalies, or neonatal management of anorectal malformations.

At this screening stage, 116 articles were excluded because they did not meet the inclusion criteria, were not directly related to anorectal malformations in neonates, did not focus on

diagnostic procedures, or were published more than 11 years ago. Articles with incomplete full text access or insufficient methodological relevance were also removed from the selection process. Ultimately, 20 articles remained that met the inclusion criteria and were subsequently used for this literature review. These selected studies provided the basis for synthesizing current evidence on clinical examination, radiological assessment, ultrasound based diagnosis, MRI evaluation, anorectal manometry, and the importance of early multidisciplinary diagnosis in neonates with anorectal malformations.

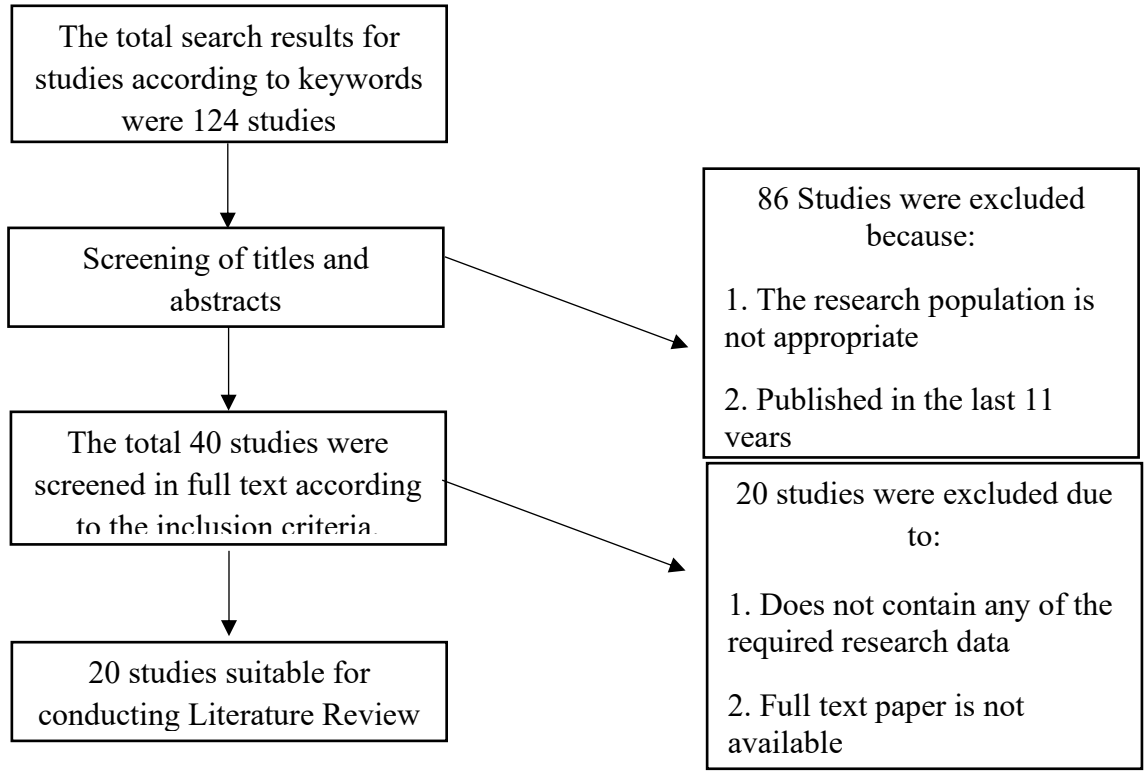

Figure 1. Flowchart of Eligible Literature

Table 1. Journal Studies on Diagnostic Approaches for Anorectal Malformations in Neonates

No	Year	Title	Method	Author	Main Findings	Conclusion
1	2022	High Resolution Transperineal Ultrasound in Anorectal Malformations. Can We Replace the Distal Colostogram?	Case series and descriptive imaging study	Palmisani et al.	High resolution transperineal ultrasound with three dimensional tomographic reconstruction was able to visualize anorectal anatomy and showed good correlation with distal colostogram findings.	Transperineal ultrasound is a promising non invasive imaging modality for preoperative ARM assessment and may reduce the need for distal colostogram in selected cases.
2	2025	High Pressure Distal Colostogram in Diagnosing Anorectal Malformations for Male Patients. Our Experience to Get a High Quality Image	Retrospective descriptive study	Ming et al.	The study summarized experience with high pressure distal colostogram in male ARM patients after colostomy and	A properly performed high pressure distal colostogram remains important for accurate fistula mapping and surgical planning

					showed that the technique helps identify fistula anatomy and improve image quality.	in male ARM patients.
3	2018	Anorectal Malformations	Clinical review	Wood & Levitt	The article reviews ARM classification, neonatal clinical evaluation, associated anomalies, surgical principles, and long term management.	Accurate early diagnosis, standardized classification, and systematic assessment of associated anomalies are essential for appropriate ARM management.
4	2015	Anorectal Malformations. A Multidisciplinary Approach	Dissertation or thesis based review	van den Hondel	The work discusses ARM as a complex congenital condition requiring multidisciplinary care, including pediatric surgery, radiology, functional assessment, psychosocial care, and long term follow up.	ARM management should not focus only on anatomical repair but should also consider continence, growth, development, quality of life, and multidisciplinary follow up.
5	2019	Contrast Enhanced Colosonography for the Evaluation of Children With an Imperforate Anus	Diagnostic case series	Chow et al.	Contrast enhanced colosonography was used to visualize preoperative anatomy in children with imperforate anus without radiation exposure.	Contrast enhanced colosonography is a useful non invasive alternative for evaluating fistula anatomy and rectal position in selected ARM patients.
6	2018	Augmented Pressure Distal Colostogram. The Most Important Diagnostic Tool for Planning Definitive Surgical Repair of Anorectal Malformations in Boys	Technical review and descriptive radiological study	Kraus, Levitt, & Peña	The augmented pressure distal colostogram provides detailed information on the distal rectal pouch and fistulous communication with the urogenital tract.	This modality remains one of the most important diagnostic tools for surgical planning in boys with imperforate anus before definitive repair.
7	2020	Image of the Month. High Pressure Distal Colostogram in a Patient with an Anorectal Malformation	Case report and image based report	Apte et al.	The report demonstrates how adequate high pressure distal colostogram technique provides essential preoperative anatomical information in male ARM patients.	Proper technique is crucial because inadequate pressure or poor imaging can lead to inaccurate anatomical interpretation before surgery.

8	2015	High Resolution MRI for Preoperative Work Up of Neonates with an Anorectal Malformation. A Direct Comparison with Distal Pressure Colostography/Fistulography	Diagnostic comparison study	Thomeer et al.	MRI was compared with distal pressure colostography and fistulography using surgical findings as the reference standard. MRI provided useful soft tissue and pelvic anatomical information.	High resolution MRI can complement or partly substitute conventional radiological evaluation, particularly in complex ARM cases and referral centers.
9	2014	Genetic and Nongenetic Etiology of Nonsyndromic Anorectal Malformations. A Systematic Review	Systematic review	Wijers et al.	The review identified genetic and non genetic factors associated with nonsyndromic ARM, although the etiology remains complex and multifactorial.	Further genetic, epidemiological, and environmental research is needed to clarify the causes of nonsyndromic ARM.
10	2022	Single Stage Procedures for Anorectal Malformations. A Systematic Review and Meta Analysis	Systematic review and meta analysis	Hartford et al.	The review analyzed published studies on single stage ARM repair and reported that selected patients may undergo single stage procedures, although surgical site infection remains an important concern.	Single stage repair may be safe in selected ARM patients, but patient selection, surgical expertise, and complication risk must be carefully considered.
11	2025	Normal Anal Sensibility in Patients Born With Anorectal Malformations	Observational clinical study	den Hollander et al.	The study showed that normal anal sensibility can still be present in patients born with different types of ARM after corrective treatment.	Sensory assessment is important for understanding functional prognosis and planning long term follow up after ARM repair.
12	2023	Diagnosing Mild Forms of Anorectal Malformation With Anorectal Manometry. A Prospective Study	Prospective diagnostic study	den Hollander et al.	Three dimensional high resolution anorectal manometry helped identify mild rectoperineal forms of congenital anorectal malformation that may be missed by routine examination.	Anorectal manometry can strengthen diagnostic evaluation in subtle ARM cases and support more accurate decisions about treatment or observation.
13	2023	Anorectal Malformations. The Pivotal Role of the Good Clinical Practice	Case report and clinical practice report	Paradiso et al.	The article highlights that ARM may be missed when newborn perineal	Careful perineal inspection and anal patency testing should be part of good

					examination is incomplete, even when the anus appears normally formed.	neonatal clinical practice to reduce missed or delayed ARM diagnosis.
14	2024	New Ultrasound Features in Diagnosing Fetal Anal Atresia. A Multicenter Prospective Cohort Study	Multicenter prospective cohort study	Kuang et al.	The study assessed new prenatal ultrasound features for detecting fetal anal atresia in a large multicenter cohort and compared their diagnostic value with traditional ultrasound signs.	New ultrasound features may improve antenatal detection of fetal anal atresia and support earlier referral, delivery planning, and postnatal confirmation.
15	2015	Anorectal Malformations	Narrative clinical review	Gangopadhyay & Pandey	The review discusses epidemiology, classification, diagnosis, and treatment of ARM, with attention to clinical management in developing country contexts.	Early detection, proper classification, and access to pediatric surgical care are essential for improving ARM outcomes, especially in resource limited settings.
16	2023	Anorectal Malformations. Ideal Surgery Timing to Reduce Incontinence and Optimize QoL	Review and clinical synthesis	Pelizzo et al.	The article discusses how surgical timing, ARM type, and individualized clinical condition influence continence outcomes and quality of life.	Timely diagnosis and carefully planned surgery are essential to optimize long term functional outcomes and quality of life.
17	2020	Analysis of Associated Anomalies in Anorectal Malformation. Major and Minor Anomalies	Retrospective cohort study	Oh et al.	The study found that associated anomalies are common in ARM and vary according to ARM subtype, including vertebral, cardiac, genitourinary, and other systemic abnormalities.	All neonates with suspected ARM should undergo systematic screening for associated anomalies to ensure comprehensive diagnosis and management.
18	2024	Late Diagnosis of Anorectal Malformation. How Good is Good Enough?	Retrospective audit and clinical analysis	Davidson et al.	The study examined delayed ARM diagnosis and identified contributing factors such as incomplete newborn examination and subtle anatomical presentations.	Standardized newborn examination, improved training, and clinical checklists are needed to reduce delayed diagnosis of ARM.
19	2024	Delayed Diagnosis of Anorectal Malformations. A	Clinical commentary and	Oyania et al.	The article discusses delayed ARM diagnosis	Standard definitions and referral

		Call for Standardization of the Current Definitions	standardization analysis		as a continuing challenge and emphasizes the lack of standardized definitions across clinical settings.	algorithms are needed to improve early recognition, reporting consistency, and quality of care for ARM patients.
20	2021	Case Report. A Case Series of Rare High Type Anorectal Malformations With Perineal Fistula. Beware of Urethral Involvement	Case series	Jun, Jacobsen, & Rai	The case series reported rare high and intermediate ARM cases with perineal fistulas that were initially thought to be low type but were later found to involve the urethra.	Clinicians should not assume that perineal fistula always indicates low type ARM. Comprehensive imaging is needed when anatomical findings are atypical.

Initial Clinical Approach and Physical Examination of the Neonate

The diagnostic approach to anorectal malformations (MAR) in neonates begins with a careful perineal clinical examination immediately after birth**.** Visual inspection for the presence of meconium, the location of the anal opening, and the direction of the perineal crease is a critical first step. Failure to detect mild abnormalities, such as a stenotic anus or small perineal fistula, is the primary cause of delayed diagnosis (Wood & Levitt, 2018). The clinical examination should also be followed by an assessment of perineal reflexes and external sphincter tone as a basis for further planning.

The Role of Conventional Radiological Examinations

The distal colostogram remains the gold standard for mapping the anatomy of the rectum and fistulas in patients with anorectal malformations who have undergone colostomy. Kraus and Apte emphasize the importance of *augmented or high-pressure distal colostogram* techniques to obtain accurate images of the rectal tip's location, its relationship with the urethra, and the type of fistula. Ming reinforces that adequate contrast pressure and proper patient positioning will result in optimal anatomical delineation without complications. However, this procedure remains invasive and involves radiation exposure, so its use is now gradually being replaced by non-invasive techniques such as ultrasound and MRI (Apte et al., 2020; Kraus et al., 2018; Ming et al., 2025).

High-Resolution Ultrasonography and Contrast-Enhanced Colosonography

Advances in ultrasound technology have transformed the diagnostic paradigm for MAR in neonates. High-resolution transperineal ultrasound (HR-US) has been shown to provide anatomical information comparable to that of a distal colostogram. This ultrasound can determine the distance from the rectum to the skin, the position of the levator ani muscle, and the direction of the fistula without the risk of radiation. Chow added that contrast-enhanced colosonography (ceCS) using microbubble contrast improves the visibility of the fistula tract and its relationship with the urethra. Kuang even demonstrated that prenatal ultrasound with detection of the absence of the "anal target sign" can aid in identifying MAR before birth. Thus, the combination of HR-US and ceCS serves as an effective, safe, and easily repeatable diagnostic tool in neonates (Chow et al., 2019; Davidson et al., 2024; Jun et al., 2021).

Magnetic Resonance Imaging (MRI) as a Complementary Modality

High-resolution MRI offers advantages in assessing soft tissue structures and complex pelvic relationships without radiation exposure. Thomeer noted that MRI has a high correlation with

distal colostograms but can provide additional information regarding sphincter muscle morphology, the sacrum, and associated pelvic anomalies. Van den Hondel & Benadering also emphasize the value of MRI for multidisciplinary surgical planning, particularly in cases of high or complex anorectal malformations. Although it requires sedation and entails higher costs, MRI is an ideal choice in referral centers with comprehensive facilities (Thomeer et al., 2015; van den Hondel et al., 2015).

Functional Approach and Manometric Examination

In addition to anatomical aspects, anorectal function is also a focus in diagnosis and further evaluation. Den Hollander et al. (2023) demonstrated that anorectal manometry can detect mild forms of MAR often missed by clinical examination, by assessing sphincter pressure and the rectal inhibitory reflex. Further studies have shown that some patients retain normal anal sensation following correction of anorectal malformations, which influences long-term prognosis. This functional evaluation is crucial for assessing surgical outcomes and determining rehabilitation needs (van den Hondel et al., 2013).

Etiological Factors and Associated Abnormalities

MAR is a multifactorial disorder involving both genetic and environmental factors. Levitt & Peña and Gangopadhyay & Pandey explain that many cases occur in conjunction with vertebral, cardiac, tracheoesophageal, and genitourinary abnormalities (the VACTERL association). Screening for associated anomalies in all neonates with suspected MAR is crucial to ensure a comprehensive diagnosis and prevent systemic complications. Knowledge of the etiology and these associations helps clinicians determine a more appropriate multidisciplinary approach. Several studies confirm that early diagnosis is closely associated with better functional outcomes, including continence and quality of life. Delayed diagnosis leads to complications such as infections, colonic dilation, and pelvic floor muscle damage. A multidisciplinary approach involving neonatologists, pediatric surgeons, radiologists, and physical therapists is key to long-term success (Kuang et al., 2024; Pelizzo et al., 2023).

Conclusion

The diagnostic approach to anorectal malformations (MAR) in neonates must be systematic and multidisciplinary, beginning with a thorough perineal clinical examination to detect abnormalities early, followed by appropriate imaging modalities based on the patient's condition. High-pressure distal colostogram remains the gold standard for determining fistula anatomy and surgical planning; however, it is now increasingly supplemented by non-invasive methods such as high-resolution transperineal ultrasound, contrast- -enhanced colonography, and high-resolution MRI, which provide detailed anatomical images without radiation exposure. Anorectal manometry also plays a crucial role in detecting mild forms of MAR and assessing postoperative anorectal function. By understanding the genetic etiology, associated abnormalities, and the importance of early diagnosis, the application of a comprehensive combination of clinical, radiological, and functional examinations can improve diagnostic accuracy, optimize surgical planning, and improve functional outcomes and quality of life for patients with MAR.

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