

## Case Series

# Use of ultrasonography in determining the type of imperforate anus: a case series

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## ABSTRACT

Preoperative differentiation between low type and high type imperforate anus (anorectal malformation, ARM) is very important to prevent the performing of inappropriate surgical procedures. Traditional invertogram is time dependent, difficult and may be harmful in neonates whose presentation is delayed and with abdominal distention. The present study assesses the diagnostic value of transperineal ultrasonography (T-PUS) in comparison with the invertogram, with the surgical diagnosis as the "gold standard", in a late-referred cohort. A prospective, interventional case series of consecutive neonates with imperforate anus seen at a tertiary referral center in the south of Iraq was performed. The typical referral was delayed (mean age >48 hours). A standard invertogram was performed (where clinically possible) and a T-PUS in the physiological supine position with a 7.5-12 MHz linear transducer for each patient. The P-P distance was recorded and the ARMs were categorized as low-type (<2.0 cm) or high-type (≥2.0 cm) ARMs. Diagnostic accuracy with respect to the intraoperative findings was the primary outcome. Secondary outcomes were feasibility and safety of each modality. Ten neonates (6 males, 4 females) were recruited. Mean age at presentation was 2.4±0.7 days (range: 1-3). In 40% (4/10) of patients, positioning the patient on the invertogram was impossible due to abdominal distension. Of the 6 patients who had both modalities, T-PUS correctly classified 10/10 (accuracy 100% (95% CI: 69.2%-100%)), while the invertogram did so in 4/6 cases (accuracy 66.7% (95% CI: 22.3%-95.7%)). In total, T-PUS was able to correctly identify 7 low-type (mean P-P distance 1.24±0.19 cm) and 3 high-type (mean P-P distance 2.63±0.15 cm) ARMs, with a 100% concordance with surgery. This difference between low and high groups was extremely significant ( $p<0.001$ , independent samples t-test). In 2 cases, the gas shadow was obscured by fluid-filled bowel loops making the invertogram unsuccessful. Transperineal ultrasonography is a safe, feasible and highly accurate tool for the diagnosis of imperforate anus, especially in cases presenting to the neonate late. This is better than the invertogram and allows for a correct and definitive surgical decision making. In all neonates with ARM we recommend that T-PUS is the primary, standalone imaging test and that the invertogram would be of no value in this setting.

**Keywords:** Anorectal malformation, Imperforate anus, Transperineal ultrasound, Invertogram, Neonate, Colostomy, Diagnostic accuracy

## INTRODUCTION

Anorectal malformations (ARMs), commonly referred to as imperforate anus, represent a complex spectrum of congenital anomalies with an estimated incidence of 1 in 4,000 to 5,000 live births.<sup>1</sup> The surgical management hinges entirely on the accurate preoperative distinction between "low-type" (translevator, infraperitoneal) and

"high-type" (suprlevator, suprapertitoneal) lesions.<sup>2</sup> A low-type ARM can be definitively managed with a primary perineal anoplasty in the neonatal period. In contrast, a high-type ARM requires a staged approach, beginning with a diverting colostomy, followed later by a definitive posterior sagittal anorectoplasty (PSARP), and finally colostomy closure.<sup>3</sup> The consequences of misclassification are profound: an erroneous perineal

dissection in a high lesion risks devastating injury to the sphincter complex and pelvic floor, leading to lifelong fecal incontinence, while an unnecessary colostomy in a low lesion subjects the infant to the morbidity of three operations instead of one.<sup>4</sup>

For decades, the standard preoperative imaging modality has been the lateral pelvic radiograph with the infant in a head-down, prone position, commonly known as the invertogram.<sup>5</sup> This technique relies on the principle that intraluminal gas rises to the most superior point of the blind rectal pouch, allowing measurement of the pouch-perineum (P-P) distance against a radiopaque marker placed on the anal dimple. The invertogram is subject to several well-characterized, critical limitations. First, the examination must be delayed for a minimum of 12-24 hours after birth to allow sufficient swallowed air to traverse the entire gastrointestinal tract and reach the rectal pouch.<sup>5,6</sup> Performing the study prior to this time window risks a falsely elevated P-P distance, misclassifying a low lesion as high. Second, the technique requires precise positioning of a distressed, often vomiting neonate in a head-down posture for several minutes, a maneuver that carries a genuine and significant risk of aspiration, respiratory compromise, and even sudden death, particularly in an infant with an already distended abdomen.<sup>7</sup> Third, the radiographic interpretation can be severely compromised in the presence of abdominal distension, where fluid-filled, dilated bowel loops adjacent to the rectal pouch obscure the gas shadow or produce a misleading summation artifact.<sup>8</sup> These limitations are magnified in low-resource or referral-center settings where patients frequently present beyond the ideal 24-hour window.

The clinical reality in our institution, a major tertiary referral center in southern Iraq, exemplifies this problem. A substantial proportion of neonates with imperforate anus are referred from peripheral hospitals or homes on day 2 or 3 of life, often with established abdominal distension, bilious vomiting, and clinical signs of intestinal obstruction. In this cohort, the invertogram is not merely an imprecise diagnostic test; it is a potentially life-threatening procedure. This acute, practical, and recurrent clinical danger prompted our department to seek a safer, equally or more accurate, diagnostic alternative.

Transperineal ultrasonography (T-PUS) has been proposed as a radiation-free, bedside-accessible, and position-independent modality for assessing the rectal pouch and sphincter anatomy.<sup>9-11</sup>

However, the existing literature consists predominantly of small case series, and none have specifically evaluated the performance of T-PUS in the uniquely challenging clinical context of delayed neonatal presentation with abdominal distension, a scenario where the invertogram is most likely to fail and be dangerous. Our study was designed to address this precise gap in the literature.

### ***Aim and hypothesis***

We hypothesized that transperineal ultrasonography would be superior to the conventional invertogram in terms of feasibility, safety, and diagnostic accuracy for distinguishing low-type from high-type imperforate anus, with intraoperative surgical findings serving as the definitive reference standard. The primary objective was to calculate and compare the diagnostic accuracy, sensitivity, and specificity of T-PUS and the invertogram. The secondary objective was to define a safe, reproducible, and evidence-based imaging protocol that could replace the invertogram as the first-line investigation in this vulnerable patient population.

### **CASE SERIES**

We present a case series of 10 consecutive, eligible neonates with imperforate anus who were enrolled and completed the study protocol. Patients were followed in a tertiary referral center in the southern part of Iraq. Ten neonates (6 males and 4 females) were selected. The mean age at presentation was  $2.4 \pm 0.7$  days (range 1-3 days). Patients had clinical evaluation, transperineal ultrasonography (T-PUS) and invertogram, if clinically feasible. Surgical reference standard was the intraoperative findings.

The T-PUS was performed in the physiological supine position using a 7.5-12 MHz linear transducer. The P-P distance was recorded and the ARMs were categorized as low-type ( $<2.0$  cm) or high-type ( $\geq 2.0$  cm).

The invertogram was performed where clinically possible. In 40% (4/10) of patients, positioning the patient on the invertogram was impossible due to abdominal distension. In two cases, the gas shadow was obscured by fluid-filled bowel loops making the invertogram unsuccessful.

The clinical, sonographic, radiographic, and surgical findings of the 10 neonates are summarized in Table 1.

T-PUS was feasible and safely completed in all 10 patients (100%). The supine position was well-tolerated by all infants, and no episodes of desaturation, vomiting, or clinical deterioration occurred during or immediately after any of the ultrasound examinations.

The conventional invertogram presented major feasibility and safety challenges. The examination was only deemed clinically safe to attempt in 6 out of 10 patients (60.0%).

In the remaining 4 patients (40.0%), the degree of abdominal distension was so severe that the surgical and radiology teams jointly decided that the head-down position posed an unacceptable risk of aspiration and respiratory compromise, and the examination was cancelled.

Of the 6 invertograms that were attempted, 2 studies were technically successful but ultimately non-diagnostic. In these two patients, the radiologist was unable to confidently identify the terminal rectal gas bubble, as it was obscured by a combination of overlying fluid-filled bowel loops and a poor, generalized gaseous ileus pattern. Therefore, a valid, interpretable invertogram measurement was obtained in only 4 out of 10 patients (40.0%).

High-quality, interpretable sonographic images were obtained in all 10 cases. Seven patients (70%) had a T-PUS P-P distance <2.0 cm and were classified as low-type ARM. The mean P-P distance in this subgroup was 1.24±0.19 cm (median: 1.20 cm; range: 1.0-1.5 cm). Three patients (30%) had a T-PUS P-P distance ≥2.0 cm and were classified as high-type ARM. The mean P-P distance in this subgroup was 2.63±0.15 cm (median: 2.60 cm; range: 2.5-2.8 cm).

The difference in the mean P-P distance between the low-type and high-type groups was highly statistically significant (1.24 cm versus 2.63 cm;  $p<0.001$ , independent samples t-test). In two of the three high-type male patients, the T-PUS successfully visualized a hypoechoic fistula tract extending anteriorly from the pouch towards the posterior urethra, suggestive of a rectourethral fistula.

Among the 4 patients with a diagnostic invertogram, the P-P distance was concordant with the clinical picture. It correctly identified 2 low-type and 2 high-type ARMs. However, as detailed above, it failed to provide a clinically useful result in the remaining 6 patients (4 cancelled due to safety, 2 non-diagnostic).

All 10 patients proceeded to surgery. The intraoperative findings served as the gold standard. All 7 patients classified as low-type by T-PUS underwent a successful primary perineal anoplasty. The rectum was found within 1.5 cm of the perineal skin in all cases, requiring no abdominal exploration. There was 100% concordance.

All 3 patients classified as high-type by T-PUS underwent a laparotomy and divided descending colostomy. Intraoperative findings confirmed a supralelevator rectal pouch in all three, with a distance from the perineum exceeding 2.5 cm. A rectourethral (bulbar) fistula was confirmed in the two male patients where it was suspected on ultrasound. There was 100% concordance.

The diagnostic accuracy of T-PUS was 100% (10/10 correct, 95% CI: 69.2%-100%). The diagnostic accuracy of the invertogram (intention-to-diagnose) was 40.0% (4/10 correct, 95% CI: 12.2%-73.8%). The difference in overall accuracy between the two modalities was statistically significant (Fisher's exact test,  $p=0.011$ ). The inter-method agreement (Cohen's kappa) between T-PUS and the diagnostic invertogram in the 4 patients where both provided a result was 1.0 (perfect agreement).

**Table 1: Clinical, T-PUS, surgical, and invertogram findings in the 10 neonates with imperforate anus.**

| Variable                                   | N (%) / value             |
|--------------------------------------------|---------------------------|
| <b>Sex</b>                                 |                           |
| Male                                       | 6 (60.0)                  |
| Female                                     | 4 (40.0)                  |
| <b>Gestational age, weeks (mean±SD)</b>    | 39.1±1.1                  |
| <b>Birth weight, kg (mean±SD)</b>          | 3.12±0.32                 |
| <b>Age at presentation, days (mean±SD)</b> | 2.4±0.7                   |
| <b>Age range (in days)</b>                 | 1-3                       |
| <b>Presenting &gt;24 hours</b>             | 9 (90.0)                  |
| <b>Abdominal distension at imaging</b>     | 9 (90.0)                  |
| <b>Bilious vomiting</b>                    | 7 (70.0)                  |
| <b>T-PUS diagnosis</b>                     |                           |
| Low-type                                   | 7 (70.0)                  |
| High-type                                  | 3 (30.0)                  |
| <b>Surgical procedure</b>                  |                           |
| Anoplasty                                  | 7 (70.0)                  |
| Colostomy                                  | 3 (30.0)                  |
| <b>Surgical correlation</b>                |                           |
| Correct                                    | 10 (100.0)                |
| <b>Invertogram feasibility</b>             |                           |
| Feasible                                   | 6 (60.0)                  |
| Not feasible (unsafe)                      | 4 (40.0)                  |
| <b>Invertogram diagnostic status</b>       |                           |
| Diagnostic                                 | 4 (40.0)                  |
| Non-diagnostic/obscured                    | 2 (20.0)                  |
| <b>T-PUS diagnostic accuracy</b>           | 100% (10/10)              |
| T-PUS sensitivity                          | 100% (95% CI: 29.2-100)   |
| T-PUS specificity                          | 100% (95% CI: 59.0-100)   |
| T-PUS PPV                                  | 100%                      |
| T-PUS NPV                                  | 100%                      |
| T-PUS overall accuracy                     | 100% (95% CI: 69.2-100)   |
| Invertogram sensitivity                    | 66.7% (95% CI: 9.4-99.2)  |
| Invertogram specificity                    | 100% (95% CI: 39.8-100)   |
| Invertogram PPV                            | 100%                      |
| Invertogram NPV                            | 50%                       |
| Invertogram overall accuracy               | 40.0% (95% CI: 12.2-73.8) |

## DISCUSSION

The central finding of this prospective case series is unambiguous: transperineal ultrasonography was a perfectly accurate, universally feasible, and completely safe diagnostic tool for differentiating low-type from high-type imperforate anus in a high-risk cohort of delayed-presentation neonates. In the same patient group, the conventional invertogram the historical gold standard failed to provide a useful clinical answer in 60% of cases,

either due to safety concerns precluding its performance or due to non-diagnostic image quality. Our results challenge the continued role of the invertogram and suggest that T-PUS should be adopted as the first-line, and likely sole, imaging modality for this condition in similar clinical settings.

Our finding of 100% diagnostic accuracy for the sonographic P-P distance is consistent with, and extends, the existing body of literature. Kim et al, in a seminal study, were among the first to demonstrate that T-PUS could reliably identify the type of ARM and internal fistulas.<sup>9</sup> Han et al using the P-P distance as the primary discriminator, reported a sensitivity and specificity exceeding 90% in a cohort of 15 patients.<sup>10</sup> More recently, a systematic review by van der Steeg et al concluded that T-PUS is a valuable tool, though they highlighted the need for more prospective data.<sup>7</sup> Our study contributes uniquely to the literature by focusing on a specific, clinically problematic subset: the neonate who arrives late, with a distended abdomen, where the diagnostic challenge is greatest. In this context, the sonographic technique is not merely an "alternative"; it is the only safe and effective option.

The failure of the invertogram in our series warrants specific discussion. The 40% rate of non-diagnostic or unsafe examinations is not an outlier but a predictable consequence of the underlying pathophysiology of advanced neonatal bowel obstruction. After 48-72 hours of obstruction, the distal bowel is typically filled with fluid and thick meconium rather than air. The air that is present is often scattered and trapped within multiple dilated loops, not concentrated in the distal rectum.<sup>12</sup> Consequently, the pathognomonic "rectal gas bubble" of the invertogram is either absent or indistinguishable from the surrounding ileus pattern. Furthermore, the mechanical risk of the head-down position is real and grave; the combination of a distended abdomen pushing against a compromised diaphragm and an incompetent gastroesophageal junction creates the perfect conditions for massive aspiration. The four cases in our series where the senior surgeon and radiologist jointly cancelled the invertogram on safety grounds underscore a critical point: a diagnostic test that carries a non-trivial risk of killing the patient has no place in modern neonatal care when a safer, more accurate alternative exists.

The transperineal ultrasound technique addresses these two critical failings directly. First, it is anatomical, not physiological: it visualizes the actual blind pouch and its contents, rather than relying on the delayed and unpredictable transit of gas. This makes it independent of the time since birth. Second, its physiological supine positioning eliminates the iatrogenic risk of aspiration. Our protocol, which is readily teachable, reduces the diagnostic evaluation to a single, objective, linear measurement that provides a clear "rule-in" or "rule-out" for a primary procedure.

### **Clinical implications and proposed algorithm**

Based on our results, we propose a simplified, evidence-based diagnostic algorithm. For any neonate presenting with imperforate anus, regardless of age or degree of distension, a bedside transperineal ultrasound should be performed immediately. If the P-P distance is  $<2.0$  cm, and the sphincter complex is well-developed, the patient can be booked for a primary neonatal anoplasty. If the P-P distance is  $\geq 2.0$  cm, a diverting colostomy is planned as the first stage. In our experience, the addition of the invertogram added no useful information and caused delay and potential harm. We have eliminated it entirely from our clinical pathway.

### **Limitations**

We acknowledge several limitations to our study. First, the sample size is small ( $n=10$ ). This is a reflection of the nature of studying a congenital anomaly in a single center. The exact binomial confidence intervals for our accuracy estimates are, consequently, wide (69-100%), and larger, multi-center studies are essential to validate these results with greater precision. Second, the T-PUS was performed by a single, highly motivated operator, and the results may be influenced by this expertise. However, the measurement we rely on is a simple linear distance, a skill fundamental to all sonography training, and we are confident it is generalizable. Third, we could not perform a randomized comparison due to the ethical constraints of assigning a patient to an examination we had already deemed clinically dangerous. The comparison is therefore based on clinical feasibility and safety, which are, in themselves, valid outcomes.

### **CONCLUSION**

Transperineal ultrasonography is a safe, feasible, and 100% accurate imaging modality for the preoperative classification of imperforate anus in neonates, particularly those presenting late with abdominal distension. It definitively outperforms the conventional invertogram, which failed in 60% of cases due to safety or diagnostic inadequacies. This simple, bedside, radiation-free test enables the surgeon to make the correct, definitive surgical decision primary anoplasty versus colostomy with complete confidence. We advocate strongly for the abandonment of the risky, time-dependent invertogram and the adoption of transperineal ultrasound as the new standard of care for the initial imaging evaluation of all neonates with imperforate anus.

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