



## Clinical Research Paper

## Beyond cloacal malformation. Addressing unclassifiable complexity

Zeni Haveliwala<sup>a,\*</sup>, Athanasios Tyraskis<sup>b</sup>, Kathryn Ford<sup>a</sup>, Tanvi Singh<sup>a</sup>, Neetu Kumar<sup>b</sup>, Hazel Learner<sup>c</sup>, Sonia Basson<sup>a</sup>, Stavros Loukogeorgakis<sup>a</sup>, Simon Blackburn<sup>a</sup>, Abraham Cherian<sup>b</sup>, Joe Curry<sup>a</sup>

<sup>a</sup> Great Ormond Street Hospital, Department of Paediatric Colorectal Surgery, Great Ormond Street, London, WC1N 3JH, UK

<sup>b</sup> Great Ormond Street Hospital, Department of Paediatric Urology, Great Ormond Street, London, WC1N 3JH, UK

<sup>c</sup> University College London, Department of Paediatric and Adolescent Gynaecology, 235 Euston Road, London, NW1 2BU, UK

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## ABSTRACT

**Introduction:** Cloacal malformations are a spectrum of complex congenital anomalies traditionally classified by common channel length, urethral length, Müllerian configuration, and sacral or spinal abnormalities. Classification guides reconstructive strategy and informs prognosis. However, a subset of patients present with atypical anatomy that defies standard classification systems, complicating surgical planning and prediction of outcomes.

**Aim:** Our cloacal malformation multidisciplinary service utilises a pathway based on common channel length, Mullerian anomalies, urethral length and sacral spinal abnormalities. We aimed to assess the management and faecal continence outcomes of these unclassifiable cases.

**Methods:** We conducted a retrospective review of 82 patients referred for primary reconstruction from 2010, analysing categorical and numerical data using Fisher's exact and Mann–Whitney U tests. Patients were categorised as classic cloaca, posterior cloaca, and unclassifiable (exhibited complex/atypical anatomy such as indistinct or absent common channels, duplication of urogenital structures or associated covered cloacal exstrophy variants).

**Results:** Of 82 patients, 15 were unclassifiable. The latter group had more co-morbidities: 75 % (n = 6) with spinal anomalies requiring neurosurgery, and 93 % (n = 14) with renal anomalies. They were more likely to undergo laparotomy (n = 10) and had poorer faecal continence outcomes, with bowel function score of 6, compared to 13 in the classic cloaca group. Surgical management was more complex, with all patients requiring functional diversion procedures rather than standard anatomical reconstruction, representing a form of “non-anatomical reconstruction”.

**Conclusion:** Unclassified cloacal malformation group represent a significant challenge, with poorer functional outcomes likely due to greater anatomical complexity and coexisting anomalies. Successful management requires experience in cloacal reconstruction and operative planning tailored to each patient's unique anatomy.

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## 1. Introduction

Cloacal malformation is among the most complex congenital anomalies encountered in paediatric surgery, with an estimated incidence of 1 in 50,000 live female births [1]. It results from an incomplete embryological partitioning of the cloaca into separate

urogenital and anorectal components, leading to a single perineal orifice that serves as the common outlet for the urinary, genital, and gastrointestinal tracts [2]. This understanding has been refuted by the Kluth [3] studies which suggest a primary defect in the development of the cloacal membrane and the changes observed are a consequence. The condition presents a broad anatomical spectrum and is frequently associated with renal, spinal, and Müllerian anomalies [4–6].

Classification systems for cloacal malformation typically rely on common channel length, with longer channels (>3 cm) associated with increased surgical complexity and less favourable functional outcomes [1]. The classification system used in our

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\* Corresponding author. Miss Zeni Haveliwala, Colorectal Fellow, Great Ormond Street Hospital, Great Ormond Street, London, WC1N 3JH, UK.

E-mail address: [zeni.haveliwala@doctors.org.uk](mailto:zeni.haveliwala@doctors.org.uk) (Z. Haveliwala).

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**Table 1**

Type (n)	Spinal anomalies
I ultraShort (IU) – CC <sup>a</sup> <1 cm	1
I short (IS) – CC 1–3 cm	23
I long (IL) – CC > 3 cm	29
II classic (posterior cloaca)	2
II variant (posterior cloaca variant <sup>b</sup> )	12
III (unclassified)	15

Classification system of cloacal malformations used at Great Ormond Street hospital.

<sup>a</sup> CC = common channel.

<sup>b</sup> Posterior cloaca variant = posterior cloacal opening with accessory perineal opening i.e. accessory urethra.

centre follows the categories shown in Table 1. However, in centres with specialist experience managing these anomalies, a subset of patients has emerged whose anatomy is too atypical to fit established categories. These unclassifiable cases present unique challenges in diagnosis, surgical planning, and prognostication.

The optimal management of cloacal malformations require a multidisciplinary approach involving colorectal surgery, urology, gynaecology, radiology, and neurosurgery [1,7]. Goals of treatment are based on achieving good functional urinary and faecal control, preserving renal function, optimising later sexual function and fertility. Despite advances in surgical techniques, long-term outcomes are variable and are influenced by associated anomalies and surgical complexity. While classification systems based on common channel length and associated abnormalities offer a useful framework, a subset of patients present with atypical or complex anatomy that defies categorisation. Unclassified cloacal malformations may exhibit overlapping features or involve multiple organ systems, complicating surgical planning and management. These complex presentations necessitate individualised treatment strategies. We believe, the clinical expertise and surgical principles developed through the management of classified cloacal malformations serve as a crucial foundation for informed decision-making in the unclassified patient group which represent a more heterogenous and challenging cohort.

Great Ormond Street Hospital is home to the largest specialist cloaca multidisciplinary cloaca clinic in the United Kingdom. The aim of this study was to review the cohort of patients referred to our cloacal malformation service who could not be assigned to an existing classification. We sought to describe their clinical characteristics, reconstructive management, and functional outcomes and to compare them to those patients who we could describe as either classic or posterior cloacal malformations.

## 2. Methods

A retrospective review was conducted of all patients with a diagnosis of cloacal malformation referred to our multidisciplinary service between January 2010 and March 2024. Patients who had reconstructive surgery, rather than only faecal or urinary diversion elsewhere, were excluded. Patients were identified through the departmental surgical database and included if they had undergone full anatomical evaluation and had available clinical and surgical outcome data.

Patients were classified according to our institutional protocol into one of the following categories: classic cloaca (subdivided into ultrashort, short, and long common channels), posterior cloaca

(classic and variant), or unclassifiable. The “unclassified” group comprised patients with atypical or complex anatomy that did not meet established classification criteria, including those with indistinct common channels or multiple system anomalies. Details are included in Table 2.

Data collected included demographic information, associated anomalies (e.g., spinal, renal, and Mullerian abnormalities), imaging findings, surgical procedures and functional bowel outcomes. Surgical data included the timing, type and approach (perineal vs. abdominal) of each intervention. We defined ‘anatomical reconstruction’ as the creation of three distinct perineal openings for the urethra, vagina, and rectum, which is considered the standard surgical goal in a typical cloacal repair. In cases where this was not possible due to anatomical complexity or comorbidity a ‘non-anatomical reconstruction’ was performed, often involving functional diversion procedures. Patients were grouped based on whether they underwent anatomical or non-anatomical reconstruction.

Bowel function was assessed using the Rintala Bowel Function Score (BFS) [8], a validated scoring system ranging from 1 to 20, incorporating seven domains: ability to hold back defecation, sensation of urge, frequency, soiling, accidents, constipation, and social impact. Each item is scored on a scale from 0 to 3, except for social problems, which is scored 0 to 2. The maximum total score is 20, with higher scores indicating better function. A score of 18–20 is considered good/excellent, 13–17 is fair, and below 13 is poor bowel function. Rintala scores were assessed in children aged over 3 years, as continence is not typically expected prior to this age in accordance with social norms around toilet training in the United Kingdom [9]. Continence data were obtained from follow-up clinic documentation, specialist nursing assessments, and standardised parent-reported outcome forms if available.

Statistical analysis was performed using GraphPad Software. (n. d.). *GraphPad Prism [Online software]* from <https://www.graphpad.com>. Categorical variables were compared using Fisher's exact test, and continuous variables were analysed with a one-tailed Mann–Whitney U test. A p-value <0.05 was considered statistically significant.

## 3. Results

### 3.1. Patient cohort and classification

Eighty-two patients referred for primary cloacal reconstruction between 2010 and March 2024 were included in this study. All patients had sufficient clinical, surgical, and outcome data available for analysis. Sixty-seven patients (82 %) were assigned to a defined cloacal subtype (classic or posterior), while 15 (18 %) were considered unclassifiable due to atypical anatomical features. The number of patients per category are summarised in Table 1. The median age at referral was 1 month in both groups (IQR 0–11 months for classifiable, 0–9 months for unclassifiable). For assessment of faecal continence outcomes, only patients aged over 3 years were included, in line with expected age for toilet training [9]. This included 9 of 15 patients in the unclassifiable group and 39 patients in the classifiable group.

### 3.2. Preoperative anomalies

Preoperative anomalies differed between the groups. Hydrocolpos at birth was more frequent in the classifiable group (50 % vs. 20 %,  $p < 0.05$ ). One third of the classifiable group required a tube vaginostomy (29 % vs. 7 %,  $p = 0.6$ ). Renal anomalies were more prevalent in unclassifiable patients (93 % vs. 50 %,  $p < 0.05$ ). Spinal anomalies were comparable between groups (53 % unclassifiable

Table 2

Patient	Anatomy	Specific Characteristics
<b>1</b> <b>Exstrophy</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Single perineal opening at base of skin fold leading to common channel Covered cloacal exstrophy variant (rectal fistula into base of bladder) BN open with associated pubic diastasis. No urethra Mullerian duplication with longitudinal vaginal septum
<b>2</b> <b>Atresia</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Posterior perineal opening leading to vaginal vault Rectum blind ending with no fistula Urethral atresia with small 'peanut-like' bladder Mullerian duplication with longitudinal vaginal septum
<b>3</b> <b>Complex</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Urethra and rectal fistula within vestibule Rectovestibular fistula Urethra with ectopic right ureter and left ectopic ureter with ureterocele entering into bladder neck. BN open Vaginal atresia and apparent Mullerian agenesis
<b>4</b> <b>Duplication</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Hemiclitoris and duplicated common channel laterally Rectal fistula opens into left sided common channel Hemibladder with urethra opening into associated common channel Hemiuterus and hemivagina opening into associated common channel
<b>5</b> <b>Exstrophy</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Single channel going into multiple blind ending pockets Covered cloacal exstrophy variant (rectal fistula into base of bladder) BN open with associated pubic diastasis Apparent right Mullerian agenesis (no right ovary) and left uterine horn
<b>6</b> <b>Complex</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Urethra and rectal fistula within vestibule Rectovestibular fistula Poorly opposed bladder neck Apparent right Mullerian agenesis, sequestered left uterine horn. Vaginal atresia
<b>7</b> <b>Complex</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Short UG sinus with blind ending pit posteriorly (vagina) and rectovestibular fistula Rectovestibular fistula Very short UG sinus Unknown. Blind ending vagina
<b>8</b> <b>Atresia</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Clitoris and mons recessed with single perineal opening Rectal atresia (orthotopic anal opening) Single common UG sinus Mullerian duplication with vaginal septum
<b>9</b> <b>Exstrophy</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Clitoris recessed with very short single common opening Covered cloacal exstrophy variant (rectal fistula into base of bladder) Short urethra with open BN and pubic diastasis Mullerian duplication with longitudinal vaginal septum
<b>10</b> <b>Duplication</b> <b>Exstrophy</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Hemiclitoris and duplicated common channel laterally Bowel plate open in lower midline (anterior abdominal wall) Short urethra from common channels laterally into hemiblasters Mullerian duplication with hemivagina and cervix seen from each common channel laterally
<b>11</b> <b>Complex</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Three openings in vestibule. Urethra patent, with remaining two openings blind ending No perineal opening. Rectal fistula seen in left hemivagina Normal urethra and bladder neck Apparent right Mullerian agenesis. Left hemivagina and hemiuterus
<b>12</b> <b>Complex</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Two openings with short UG sinus Unable to assess (conjoined twin) Normal urethra Mullerian duplication with longitudinal vaginal septum
<b>13</b> <b>Atresia</b> <b>Duplication</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Single common channel leading to blind ending pouch Two small openings at base of common channel (likely appendices) No urethra or bladder neck. Two hemiblasters. Hemiureters and hemivaginas which communicate with common channel in midline
<b>14</b> <b>Atresia</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Two perineal openings side-by-side in vestibule leading to single vagina No rectal fistula (blind ending) Urethra opens into vagina anteriorly Single Mullerian structure with vaginal septum distally (opens into common cavity proximally)
<b>15</b> <b>Complex</b>	<b>Perineal</b> <b>Anorectal</b> <b>Urological</b> <b>Mullerian</b>	Two perineal openings in vestibule Rectovestibular fistula Bladder opens directly into introitus. No urethra. Vaginal atresia with no perineal opening. Atretic vagina seen behind bladder

Individual anatomical clinical profile of patients in the unclassified malformation group.

BN = bladder neck, UG = urogenital.

vs. 48 % classifiable,  $p = 0.7$ ), but the need for neurosurgical intervention was significantly higher in the unclassifiable cohort (75 % vs. 21 %,  $p < 0.05$ ). Müllerian anomalies occurred more frequently in unclassifiable patients (86 % vs. 61 %), although this did not reach statistical significance ( $p = 0.1$ ).

### 3.3. Clinical characteristics

This subgroup of 15 patients demonstrated a spectrum of atypical and complex cloacal variants that defied conventional

classification. A feature across many patients was the presence of complex urogenital and anorectal anomalies, often involving short or absent common channels, posterior perineal or single perineal openings, and atresia of one or more structures (e.g., urethral, rectal, or vaginal atresia). The common channel commonly led to blind ending pouches. Pubic diastasis was observed in just under half the cohort, in association with covered cloacal exstrophy variants or midline abdominal wall defects. One patient had significant gastroschisis with associated caudal duplication (two hemiblasters, hemiuteri and hemivaginas) and duplicated common

channels of which one contained the rectal fistula. Several patients had Müllerian anomalies, including vaginal duplication, agenesis or unilateral uterine remnants. The bladder neck was often severely compromised, with a wide-open configuration in five patients, leading to the need for bladder neck closure and the formation of a catheterisable channel for bladder emptying.

In many patients, standardised reconstructive pathways could not be applied, and several children were managed with end stomas or urinary diversion alone due to anatomical limitations. For example, patients with duplicated hemibladders, or significant pelvic anomalies required staged or non-anatomical management. The heterogeneity in anatomy and multisystem involvement highlighted the surgical complexity of this group and the need for individualised management strategies (Table 2).

### 3.4. Surgical management

The median age at reconstructive surgery was similar between groups (12 months, IQR 7–23 vs. 11 months, IQR 6–19). At the time of review 14 patients in the unclassifiable group had sufficient operative data for analysis, compared with 64 in the classifiable patient cohort. The intention of treatment was to enable patients to obtain control of urine and stool whilst preserving renal function. Laparotomy was significantly more common in the unclassifiable group (71 % vs. 36 %,  $p < 0.05$ ). Two unclassifiable patients did not undergo laparotomy for reconstruction; one had covered cloacal exstrophy and a lipomyelomeningocele and opted to remain with an end ileostomy but required a laparoscopic-assisted ureterostomy; the second had a very short common channel and absent vagina that underwent a posterior sagittal anorectoplasty alone.

Bladder neck closure was considered in patients with no or minimal potential for urinary continence due to their anomalies (e.g. absent or short urethra with wide bladder neck opening into the

common channel, typically seen more commonly in the long common channel cloacas). Bladder neck closure or urinary diversion was performed more frequently in the unclassifiable group (35 % vs. 14 %), although this difference was not statistically significant ( $p = 0.1$ ).

The requirement for additional surgical intervention following initial reconstruction was comparable (35 % vs. 44 %,  $p = 0.6$ ). The need for further procedures most commonly included dilatation of the neanus, anoplasty and mitrofanoffscopy or suprapubic catheterisation for urinary drainage.

Importantly, a significantly greater proportion of children in the unclassifiable group had undergone a 'non-anatomical reconstruction' at the time of review (14/14, 100 %) compared to the classifiable group (11/63,  $p < 0.05$ ). A summary of patient classification, associated anomalies, surgical details, and faecal continence outcomes is provided in Table 3.

### 3.5. Functional bowel outcomes

Bowel function, assessed using the Rintala Bowel Function Score (BFS) in children over 3 years of age, was significantly poorer in the unclassifiable group (median BFS 6, IQR 5–7) compared to the classifiable group (median BFS 13, IQR 8–17;  $p < 0.05$ ). These differences in bowel function are illustrated in the box-and-whisker plot (Fig. 1). Persistent stoma was observed in 4 unclassifiable patients and 3 classifiable patients ( $p < 0.05$ ).

## 4. Discussion

Cloacal malformation represent a rare and heterogeneous group of congenital anomalies. Within this spectrum, the unclassifiable variants pose complex clinical and surgical challenges.

**Table 3**  
(Summary table of patient cohorts, pre-operative anomalies, surgical intervention and faecal continence outcomes) with comparison between classified and unclassified cases

	Classifiable n (%)	Unclassifiable n (%)	<i>P value</i>
Number of patients	67	15	
Median age at referral (m)	1	1	
IQR <sup>a</sup>	0–11	0–9	
Pre-operative anomalies			
Hydrocolpos at birth	34 (50)	3 (20)	<b>&lt;0.05</b>
Tube vaginostomy	20 (29)	1 (7)	0.6
Spinal	32 (48)	8 (53)	0.7
- Need for neurosurgical intervention	7 (21)	6 (75)	<b>&lt;0.05</b>
Renal	34 (50)	14 (93)	<b>&lt;0.05</b>
Mullerian	41 (61)	13 (86)	0.1
Surgical intervention <sup>b</sup>			
Age at reconstructive surgery (m)	11	12	
IQR <sup>a</sup>	6–19	7–23	
Need for bladder neck closure and urinary diversion	9 (14)	5 (35)	0.1
Laparotomy required	23 (36)	10 (71)	<b>&lt;0.05</b>
Non-anatomical reconstruction	11 (17)	14 (100)	<b>&lt;0.05</b>
Need for further surgery	28 (44)	5 (35)	0.6
Outcomes			
Median Rintala score (children >3 years of age) <sup>c</sup>	13	6	<b>&lt;0.05</b>
IQR <sup>a</sup>	8–17	5–7	
Persistent stoma (children >3 years of age) <sup>c</sup>	3	4	<b>&lt;0.05</b>

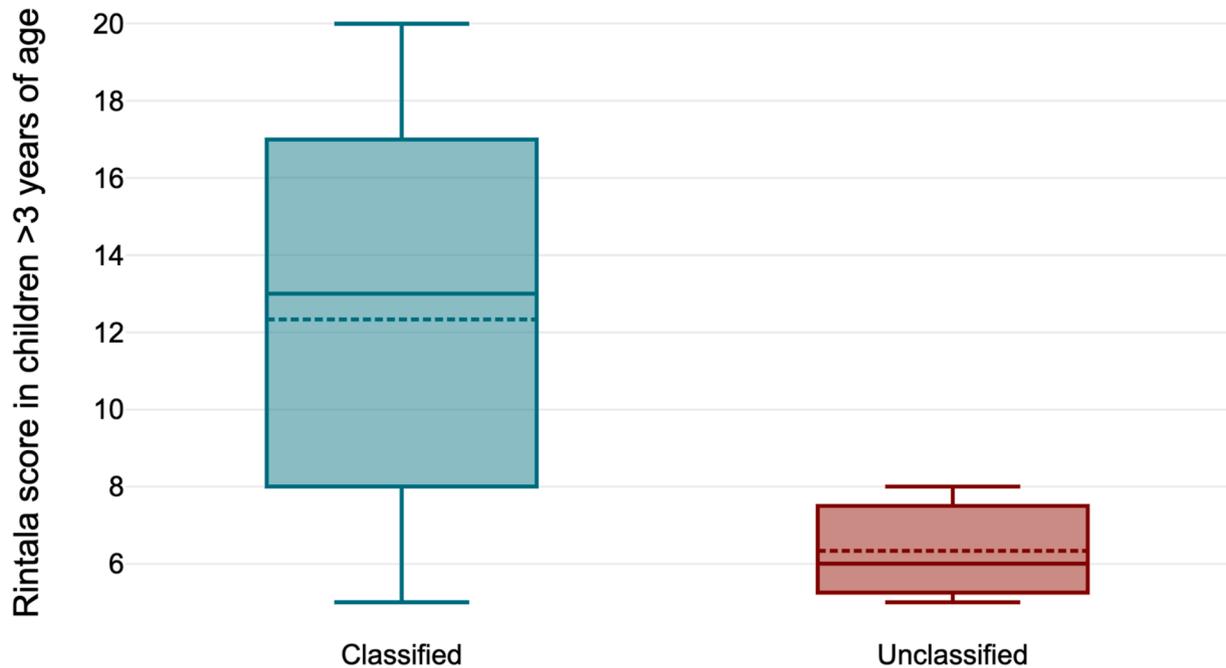
Summary table of patient cohorts, pre-operative anomalies, surgical intervention and faecal continence outcomes with comparison between classified and unclassified cases.

The bold italic numbers in the table represent significance. I.e.  $p < 0.05$  is statistically significant.

<sup>a</sup> IQR = interquartile range.

<sup>b</sup> Classifiable patients = 64, unclassifiable patients = 14.

<sup>c</sup> Children >3 years of age: classifiable patients = 39, unclassifiable patients = 9.



**Fig. 1.** Box-and-whisker plot showing the distribution of BFS for patients >3 years of age in both classifiable (n = 39) and unclassifiable (n = 9) groups. The median BFS was significantly lower in the unclassifiable group (median 6, IQR 5–7) compared to the classifiable group (median 13, IQR 8–17) ( $p < 0.05$ , Mann–Whitney U test).

Even with over 15 years of multidisciplinary experience in managing complex cloacal malformations, this subset of patients has challenged our service to provide nuanced and individualised care.

Our series highlights a distinct cohort of patients with cloacal malformations who do not fit existing classification systems. These unclassifiable patients demonstrate anatomical complexity, with features such as vaginal or urethral atresia, common channels with multiple blind pouches, and associations with other major congenital anomalies including cloacal exstrophy variants and pubic diastasis. These children present with more associated anomalies, including spinal dysraphism requiring neurosurgical intervention and renal tract abnormalities.

The higher proportion of patients in the unclassifiable group requiring non-anatomical reconstruction reflects the degree of anatomical complexity in this patient cohort. It emphasises the importance of multidisciplinary planning and setting realistic expectations with families. These decisions are not made lightly – they are guided by intraoperative findings and clinical judgement regarding long-term outcomes. In several cases, diversion was chosen to preserve renal function and optimise quality of life, particularly when there is a low likelihood of achieving continence through anatomical reconstruction. This emphasises the need to recognise unclassifiable patients as a distinct group requiring individualised surgical strategies.

We found a significantly lower Rintala Bowel Function Scores (BFS) in this group, which likely reflects the anatomical complexity, but also the observation of spinal anomalies, particularly those requiring treatment and the impact on sacral nerve function.

While traditional cloacal malformations are classified based on common channel length and associated anatomy [10,11], our findings highlight the limitations of these frameworks when applied to patients with unique abnormalities. Atypical presentations have been sparsely described in the literature, primarily in case reports or small series [12–15]. The heterogeneity in our cohort suggests that such anomalies may be under-recognised or misclassified.

These findings support individualised management plans and early multi-disciplinary team involvement, including colorectal surgery, urology, gynaecology and psychological support. The high incidence of renal and spinal anomalies in the unclassifiable group, many of which required neurosurgical intervention suggests that these patients may lie at the more severe end of the VACTERL spectrum. This further highlights the complexity of their condition and the need for integrated, coordinated care to optimise outcomes.

Counselling families and young people with unclassifiable cloacal malformations about future sexual and reproductive outcomes is challenging. This is a consequence of the limited numbers and heterogenous nature of this population. Furthermore, the time required for meaningful outcome assessment necessitates long-term follow up into adulthood. As such, current prognostication is limited, and clinicians must approach these discussions with caution, transparency, and a focus on individualised care planning.

A key strength of our study is the use of standardised outcome measures, such as the Rintala bowel function score, to assess functional outcomes across age-appropriate patients. However, limitations include the retrospective nature of the study, and the relatively small sample size of the unclassifiable group. Each patient presented with complex anatomical challenges which make broad generalisations difficult. The assessment of spinal abnormalities remains challenging, as imaging findings do not always correlate with clinical outcomes. We observed a significantly higher rate of neurosurgical intervention among the unclassifiable cohort, which may reflect selection bias. Patients who are offered surgery are often those in whom intervention is expected to improve function, rather than those with the most severe underlying pathology. Therefore, neurosurgical treatment does not necessarily indicate a more significant spinal anomaly, and its relationship with continence outcomes is difficult to quantify. A prospective, standardised evaluation of spinal and sacral anatomy, coupled with long-term functional follow-up, would be better to understand these associations.

Future work should aim to expand anatomical classifications to account for these complex anomalies and develop consensus on optimal surgical strategies.

This cohort's complexity emphasises the importance of centralised care in high-volume centres with established cloaca pathways and specialist multidisciplinary clinics. The volume and diversity of cases seen at our centre has informed our operative strategies. As these children transition to adolescence and adulthood, ongoing collaborative care will be key to supporting their medical, functional, and psychosocial needs.

In conclusion, our cohort recognises a subset of malformations that do not easily fit into a clear classification system. All patients in this study were managed through a multidisciplinary team approach. However, the unclassifiable group required more tailored planning, with greater reliance on team-based decision-making and clinical judgement. The experience of our multidisciplinary team, built over many years managing complex cloacal anomalies, was key to navigating these challenges.

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