Bowel Function in Children with Low Anorectal Malformations after Surgical Repair

A Retrospective Single-Center Cohort Study

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Keywords

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Abstract

Objective

Bowel function in patients post-surgical repair of low forms of anorectal malformations (ARMs) was evaluated. This study aimed to identify predictors of functional outcomes to guide parental counseling and predict quality of life.

Methods

Patients treated for ARMs at Ghent University Hospital between 2005 and 2015 were retrospectively analyzed. Data included demographics, ARM type, diagnosis timing, operative management, and associated anomalies. Bowel function was assessed using both Rintala questionnaire filled in by parents, resulting in bowel function score (BFS), and clinical outcome (CO) evaluated by the surgeon.

Results

In total, 80 patients were analyzed, of which 59 girls (74%), with a median follow-up age of 7.7 years. Early diagnosis (within the first week of life) occurred in 61%. The most common ARM type was rectoperineal fistula (87.5%). Associated anomalies were present in 40% of patients, with 12.5% having VACTERL association. Normal BFS (≥18/20) was achieved in 47.5% of patients, with 54% having excellent CO. There was a significant correlation between BFS and CO ($p<0.001$). Rectal trimming was associated with lower BFS ($p=0.003$). Presence of a developmental disorder significantly impaired BFS ($p=0.013$). No significant differences were observed based on timing of diagnosis or surgical intervention.

Conclusion

Half of the patients achieved excellent BFS and CO at mid-long follow-up, with significant negative impact of rectal trimming during surgery and presence of a developmental disorder later in life. Literature on trimming is absent, however our data suggests significant importance on BFS. Rintala questionnaire correlated well with CO, suggesting being useful in follow-up.

Introduction

Anorectal malformations (ARMs) represent congenital anomalies affecting the anorectal region, exhibiting a spectrum of defect severity (1). During embryonic development, abnormal cloacal division may lead to the manifestation of ARMs, although the precise etiology remains unexplained (2). It occurs in approximately 1 to 5,000 live births (3). Classification systems such as the early Wingspread or Peña classifications, and commonly used Krickenbeck classification from 2005 have been developed in an attempt to categorize ARM types based on anatomical features, although their therapeutic and prognostic utility may be limited (1). The diagnosis of ARMs typically occurs postnatally at birth through careful clinical examination, however delay with diagnosis later in life is possible when children present with functional complaints such as constipation or fecal incontinence. Correct diagnosis is essential for appropriate management and favorable patient outcomes (1, 4). ARMs may occur in isolation or in association with other congenital anomalies, with the VACTERL association being the most common (3). Surgical intervention is typically required for ARM management, with posterior sagittal anorectoplasty (PSARP) remaining the gold standard for definitive repair for low forms of ARM (5, 6). The choice between single-stage definitive repair and staged procedures involving colostomy creation depends on the type and complexity of ARMs (7). The fistulous tract is generally trimmed during surgery; however, informal debate among surgeons and sparse literature question whether this segment should be retained as it may play a role in sensitivity and continence (8-10). Long-term follow-up is essential to monitor bowel function, including, soiling, fecal incontinence or constipation (11). In pediatric patients with ARMs, incontinence

can result from an inadequate sphincter mechanism or reduced bowel sensitivity. Additionally, prolonged bowel distention may decrease bowel motility, which can exacerbate tendencies towards constipation. Both mechanisms underlying incontinence may coexist, compounding their effects (7). However, prognosis in patients after ARM repair remains multifactorial, influenced by anatomical complexity, associated anomalies, and treatment approach, including timing and approach of surgery and the integrity of neural and muscular structures. For parents to be properly informed about the child's prognosis, it is essential to accurately assess the likelihood of bowel dysfunction including incontinence, soiling and/or constipation later in life. However, there's no consensus in literature regarding the demographic and clinical characteristics that may have a prognostic value.

Objective

This study aimed to evaluate bowel function after surgical repair of low forms of ARMs in a single center cohort, exploring the relation between bowel function at mid-long follow-up, and demographic, clinical, and therapeutic characteristics.

Materials and methods

Study Population

This retrospective study received approval from the Ghent University Hospital ethics committee, Ghent, Belgium (No. BE670201837662). This single center study (Ghent, University Hospital, Ghent, Belgium) included patients treated or operated on between January 2005 and December 2015 for low forms of ARM including anal stenosis, rectoperineal fistula, rectovestibular fistula, and imperforate anus without fistula.

Data Collection

The treating physicians contacted parents or legal guardians by phone to request participation, and written informed consent was obtained. Rintala questionnaires were mailed to participants and returned by post. Simultaneously, demographic and clinical data were retrospectively collected from medical records, including ARM type, timing of diagnosis, presenting symptoms, preoperative interventions, surgical details, postoperative complications, and associated anomalies. Early diagnosis was defined as within the first week of life.

Questionnaire

The Rintala questionnaire, consisting of seven questions with corresponding scores, was used to assess bowel function (12). Questions included information concerning control of defecation, stool urge, stool frequency, soiling, incontinence, constipation, and social hindrance/discomfort in the context of bowel function. Each answer on the questionnaire corresponded to a score, with a global score known as the bowel function score (BFS) based on the responses. The maximus BFS that could be obtained was 20, where a BFS of at least 18 out of 20 was considered indicative of normal bowel function (12).

Scoring of function by expert opinion

The Clinical Outcome (CO) was assessed simultaneously by the treating surgeon. Information was derived from the last medical report at follow-up in policlinics, where outcomes were categorized as excellent, good, moderate, or poor, based on criteria related to bowel movements, social limitations, and constipation severity. CO as evaluated by the treating surgeon and BFS as reported by the patient's parents was obtained independently from each other. Both were used to provide a comprehensive mid-term view of the patient's bowel function.

Statistical Analyses

Statistical analysis was performed using SPSS Statistics, version 26 (IBM Corp., Armonk, NY, USA). Descriptive statistics were used to assess the distribution of the BFS, seen as a continuous, independent variable. We analyzed the association between BFS and predefined factors with potential influence, including the timing of surgery, type of surgery (with particular attention to trimming of the anorectum), and the presence of associated sacral/spinal anomalies. Mann-Whitney U test and the Kruskal-Wallis test were conducted due to the non-parametric distribution of the data. Based on the results of the bivariate analyses and findings from the literature, several potential variables were further analyzed as predictors using multivariate analysis in the form of multiple linear regression. The correlation between BFS and CO was evaluated using the Spearman rank correlation test. All statistical tests were two-sided, with a significance level set at $p < 0.05$.

Results

Study Population

Of the 110 patients diagnosed with a low form of ARM within the study period, two were excluded due to death from unrelated causes. Consent was obtained from the parents or legal guardians of 85 patients. Ultimately, a dataset of 80 patients was analyzed

TABLE 1: Low forms of Anorectal Malformations Types.

	Rectoperineal fistula	Rectovestibular fistula	Imperforate Anus	Total n (%)
Total N (%)	70 (87,5%)	9 (11,3%)	1 (1,3%)	80 (100%)
Gender N (%)				
Girls	50 (71,4%)	9 (100%)	0 (0%)	59 (73,7%)
Boys	20 (28,6%%)	-	1 (100%)	21 (26,3%)
Trimming, N (%)				
Yes	45 (64,3%)	4 (44,4%)	0 (0%)	49 (61,2%)
No	19 (27,1%)	4 (44,4%)	1 (100%)	24 (30%)
unknown	6 (8,6%)	1 (11,2%)	0 (0%)	7 (8,8%)

TABLE 2: Associated anomalies diagnosed in patients with low Anorectal Malformations at Ghent University Hospital.

System	Total N (%)	Associated anomalies
Cardiac	22 (27,5)	Atrial septal defect; Ventricular septal defect; Tetralogy of Fallot; Persistent ductus arteriosus; Patent foramen ovale; Pulmonary valve stenosis
Renal/Urogenital	11 (13,8)	Renal dysplasia; Vesicoureteral reflux; Hemi-uterus; Neurogenic bladder; Hypospadias; Solitary/Ectopically implanted/Multicystic kidney
Cranial	11 (13,8)	Facial dysmorphia; Preauricular tag; Malformation of the auricle; Microcephaly; Microretrognathia; Constricted ear
Limb	9 (11,3)	Clubfoot; Thumb appendages; Thumb hypo-/aplasia, Radial aplasia
Spinal/vertebral	6 (7,5)	Vertebral fusion; Missing/additional vertebrae; Tethered cord
Gastro-intestinal	2 (2,5)	Esophageal atresia with tracheoesophageal fistula; malrotation of the duodenum

after excluding patients due to conservative treatment (2), treatment abroad due to complexity of surgery (associated vaginal agenesis) (1), or incomplete questionnaires (2).

Out of the 80 included patients, 59 were girls (74%). The majority of patients (80%) were born after 37 weeks gestational age. Of the 8 (10%) premature infants, the median gestational age was 35 weeks (range: 28- 36 weeks), with unavailable data on gestational age in the remaining 8 patients. Diagnosis occurred early within 48 hours after birth in 54% of neonates (43/80), with an additional 7% within 7 days of life (49/80). In the remaining 31 children where diagnosis was initially missed in the first week of life, one or more associated symptoms which led to diagnosis included obstruction (8/31), constipation (31/31), incontinence (1/31) and painful defecation (11/31).

TABLE 3: Rintala Questionnaire: Evaluation of bowel function.

System	Total N (%)	Associated anomalies
Factors	Score given	N (%)
Total filled in questionnaires		80 (100%)
Ability to hold back defecation		
Always	3	49 (61%)
Problems less than 1/week	2	17 (21%)
Weekly problems	1	7 (9%)
No Voluntary Control	0	7 (9%)
Feels/reports the urge to defecate		
Always	3	50 (62,5%)
Most of the time	2	18 (22,5%)
Uncertain	1	8 (10%)
Absent	0	4 (5%)
Frequency of Defecation		
Every other day - twice a day	2	64 (80%)
More often	1	9 (11%)
Less often	0	7 (9%)
Soiling		
Never	3	12 (15%)
Staining less than 1/week, no changer of underwear required	2	37 (46%)
Frequent Staining/soiling, change of underwear often required	1	19 (24%)
Daily soiling, requires protective aids	0	12 (15%)
Accidents		
Never	3	45 (56%)
Less than 1/week	2	22 (27%)
Weekly accidents, requires protective aids	1	6 (8%)
Daily, requires protective aids during day and night	0	7 (9%)
Constipation		
No Constipation	3	53 (66%)
Manageable with diet	2	8 (10%)
Manageable with laxatives	1	14 (18%)
Manageable with enemas	0	5 (6%)
Social problems		
No social problems	3	67 (84%)
Sometimes (foul odors)	2	7 (9%)
Problems causing restriction in social life	1	5 (6%)
Severe social and/or psychic problems	0	1 (1%)

Anorectal Malformation types and Associated Anomalies

Data is presented in Table 1 and Table 2. A rectoperineal fistula was present in 70 (88%) patients, a rectovestibular fistula in 9 (11%) girls, and an imperforate anus in 1 (1%) boy. A rectoperineal fistula was the most frequent type in both boys and girls: 20/21 (95%) and 50/59 (85%) respectively. Associated congenital anomalies were seen in 32 (40%) patients. A VACTERL association, defined as at least 3 out of 7 systems affected, was subsequently seen in 10 of these 32 children (12.5% out of the total population). Additionally, out of 80 patients, 13 (16%) children exhibited chromosomal anomalies.

Preoperative, Operative, and Postoperative Data

Preoperative management included dilatation in 14 (17%), irrigations in 9 (11%), and use of laxatives in 22 patients (27%). All patients underwent PSARP for definitive repair. Sixty-eight patients (85%) had single-staged repair, whereas repair after an initial colostomy creation was seen in 12 patients (15%). The median age at definitive repair was 76 (IQR 38 – 150) days, with a total range from day 1 of life to 9,3 years old. When comparing the age at definite repair for patients with single-staged repair or with colostomy creation, respectively, a median age of 61 (IQR 31 – 150) days versus 108 (IQR 73 – 156) days was seen. The fistulous tract was trimmed in 49 patients (61%) (Table 2). Postoperative complications occurred in 21 (26%) patients, including urological co-morbidities in 3 (4%), anal mucosal ectropion in 11 (14%), and wound dehiscence in 7 (9%) patients. There were 0 cases of anal stenosis.

Rintala Questionnaire

Data is presented in Table 3. All parents completed the Rintala questionnaire. The median age of patients at the time of completion of the questionnaire was 7.7 (IQR 5,6 – 10,1) years. Median BFS was 17 (range 3 - 20), with 38 out of 80 patients (47.5%) with a BFS of at least 18, which correlated with a normal bowel function.

Urge to defecate was evaluated with most children (62.5%) always feeling the urge to defecate, and 22.5% who did most of the time. However, 10% of parents were unsure about their child's awareness, and 5% reported that their child did not feel the urge to defecate.

Regarding bowel accidents, 56% of children didn't experience any, with 27.5% having infrequently bowel accidents. In 7.5% of cases weekly problems were reported. Nine percent of children needed diapers day and night due to daily accidents.

A significant number of children had some degree of soiling, with only 15% never experiencing it. Nearly half (46%) had soiling less than once a week, however, 23% soiled weekly, necessitating frequent changes of underwear. A total of 15% experienced daily soiling, requiring additional protective measures such as pads or diapers.

Most children (66%) did not suffer from constipation. Dietary measures were sufficient to manage constipation in 10% of cases, while 17.5% required laxatives. A small percentage (6%) needed more intensive interventions such as enemas.

Social problems were reported absent in almost all children (84%). The remaining children did report occasional issues, such as odor problems (9%), and 6% experiencing limitations in social interactions and activities. Only 1 child (1%) faced severe social or psychological issues.

TABLE 4: Relationship between clinical Outcome and Bowel Function Score.

Clinical Outcome	N (%)	BFS-score Median (range)	BFS ≥ 18 - N (%)	BFS < 18 - N (%)
Excellent	40 (54%)	18 (11 - 20)	27 (67,5)	13 (32,5)
Good	20 (27%)	16,5 (3 - 20)	9 (45)	11 (55)
Moderate	5 (7%)	14 (8 - 16)	0 (0)	5 (100)
Poor	9 (12%)	7 (3 - 14)	0 (0)	9 (100)
Total	74		36 (49)	38 (51)

TABLE 5: Predictors of Bowel Function Score.

		Bowel Function Score			
Variable	Total (n)	Median	IQR	Range	P-value
Gender					
Girls	59	17	14 – 19	3 - 20	0,791
Boys	21	16	14 – 19	7 - 20	
ARM-type					
Rectoperineal	70	17	14,75 – 19	3 – 20	0,166
Rectovestibular	9	15	7,5 – 18,5	4 – 19	
Gestational age					
Term	64	17	14 – 19	3 – 20	0,138
Preterm	8	14,5	8 – 17,5	3 – 20	
Timing of diagnosis					
Early (first week of life)	49	17	12,5 – 19	3 – 20	0,576
Late (after first week of life)	31	18	15 – 19	4 – 20	
Timing of surgery					
< 1 week	13	17	12,5 – 19,5	3 – 20	0,368
1 week - 4 months	41	18	14 – 19	3 – 20	
> 4 months	26	16	14,75 – 18	4 – 19	
Sacral/spinal anomalies					
Yes	6	13	7,75 – 16,75	7 – 19	0,086
No	74	17,5	14,75 – 19	3 – 20	
Development disorders					
Yes	13	11	5,5 – 18	3 – 20	0,013
No	67	17	15 – 19	3 – 20	
Trimming					
Yes	49	16	14 – 18	3 – 20	0,003
No	24	19	16,3 – 19	8 – 20	

Clinical Outcome

Data is presented in Table 4. CO was assessed by the treating surgeon, based on the last follow-up consultation, with a total of 74 assessments, as six patients had a follow-up in another center. They did complete the Rintala Questionnaire. The median age at determination of CO was 4 (IQR 3 – 5,5) years. The median time of follow-up in policlinics was 43 (IQR 32; 63) months. In total 40 (54%) patients had excellent CO, 20 (27%) had good CO, 5 (7%) had moderate CO and 9 (12%) had poor CO. CO was significantly correlated with BFS (p <0.001). Table 4 gives an overview of the correlating BFS within each clinical group, and the percentage of patients with a BFS with a normal (≥ 18/20 BFS) or abnormal (<18/20 BFS) bowel function.

Predictors of functional outcome

Data is presented in table 5. Bivariate analyses was used to assess several variables potentially influencing functional outcome. Trimming of the rectum resulted in a significant lower BFS (median BFS 16; IQR 14 - 18) compared to patients without rectum trimming (median BFS score 19; IQR 16,3 - 18) (p = 0,003).Additionally, when evaluating rectoperineal fistula only, patients with a non-trimmed approach (n = 19; median BFS 19, range 4 - 20) scored significantly better than those with a trimmed rectum (n = 45; median BFS 16; range 3 - 20) (p = 0,004). Moreover, the presence of a developmental disorder was associated with a poorer prognosis as well in terms of BFS, with a median score of 11 (IQR 5,5 – 18), in comparison without developmental disorder (median 17; IQR 15 – 17) (p = 0,013). The timing of diagnosis nor timing of intervention had any effect on BFS (p = 0,576 and p = 0.368, respectively).

The multivariate analysis confirms results of the bivariate analysis, indicating that rectum trimming (p = 0.004) and the presence of a developmental disorder (p = 0.001) having a significant effect on bowel function. Non-significant variables included spinal anomalies (p = 0.164) and the type of anorectal malformation (ARM) (p = 0.15).

Discussion

ARMs represent a frequent congenital anomaly in children where correct diagnosis, appropriate surgical management, and long-term follow-up are crucial for optimal bowel function later in life.

Timing of diagnosis and surgery

ARMs are typically identified at birth, where diagnosis depends heavily on careful clinical observation and detailed inspection of the perineal area. Early diagnosis is defined in the literature within the first 48 hours of life, or by extension the first week of life (13-15). Jonker et al. found that complex ARMs were diagnosed early in 100% of cases, while only 54% of anatomically less complex ARMs were diagnosed early (13). In this study, more than one-third were not diagnosed within one week. This may be explained as only low forms of ARM were included, mimicking normal anatomy during the postnatal period (15). Early recognition is essential, as delayed diagnosis is associated with more preoperative complications, including severe abdominal distention (69% vs. 43% in early diagnosis) and sepsis (38% vs. 21%), as represented in a prospective cohort study by Reddy et al. (14). They also reported higher mortality rates in the delayed diagnosis group (4 out of 54 neonates, compared to 0 in the early diagnosis group), all attributed to sepsis, although this was not statistically significant. When looking at mid-term follow-up they showed no significant differences, with similar results of BFS obtained in children independent of timing of diagnosis.

Timing of surgery for ARMs is widely debated. Early neonatal surgery may have positive effects due to early relief of intestinal subobstruction and earlier acquisition of a physiological defecation mechanism. The median time for definitive repair in this cohort was 76 days, ranging from the first day of life until 9 years of age. Peña et al. (7, 16), who introduced the PSARP technique in 1980, recommended definitive repair within the first two months of life. Subsequent studies have compared the timing of surgery with post-operative complications and long-term outcomes. Pelizzo et al. used the Rintala Questionnaire and found that early surgery within 3 months correlated with better colonic function scores (> 18), although not statistically significant (17). Harumatsu et al. showed significant differences in overall BFS at the age of 11 years between early (before 5 months) and late (after 5 months) surgical repair groups, with better constipation scores in the early surgery group over time (18). Other parameters like incontinence, soiling, and bowel movements showed no significant differences, and neither was scoring in younger age groups. Harumatsu et al. only included intermediate to high types of ARM, complicating direct comparisons (18).

Type of surgery

PSARP was used in all cases, reflecting a homogeneous surgical management. A colostomy was required in 15% of patients before definitive repair. In comparison, a recent large cohort study over the UK and Ireland by Long et al. reported a high number of colostomies before or during definitive repair, present in 74% of the total population, with an incidence of 37% of those with perineal fistula and 78% in those with vestibular fistula (15). The higher incidence rates were explained by local habitude (3 stage approach), need for emergency decompression and context-related factors such as prematurity or other associated anomalies. Single-stage repairs are generally associated with better prognoses compared to staged procedures (19). However, a recent systematic review by Hartford et al. compared single-stage and staged repairs, finding no evidence of differences in long-term functional outcomes regarding voluntary bowel movements, soiling, and constipation between the two approaches (20). When looking at outcome later in life, Lauriti et al. conducted a systematic review on single-stage repair in females with rectovestibular fistula, showing no association between a one-stage approach and increased fecal incontinence (21). Single-staged repair is preferable as it minimizes the morbidity of colostomy, and need for multiple procedures under general anesthesia, however does not change long-term functional outcomes.

During the PSARP technique, the distal rectum, including the ectopic anal canal or fistula tract, is generally resected or "trimmed"(16). This topic is rarely addressed in the literature and is primarily discussed informally among pediatric surgeons (e.g., at congress meetings), with no consensus on whether to retain this segment. Trimming is typically necessary in cases of stenosis or damage (e.g., rectal atresia) (8). However, a recent study by Hamrick et al. investigated a preservation approach in fourteen patients with rectal atresia and three with rectal stenosis, describing a technique to spare the anterior dentate line (8). For other forms of anorectal malformations (ARM), opinions diverge. Some surgeons argue that the distal rectum does contain a dentate line, essential for sensitivity and continence, whereas it was previously assumed that this segment was poorly developed or even absent, favoring resection (8, 9). The ectopic anal canal was often reclassified as a fistula, leading to its routine destruction without scientific justification (16). Levin is among the few authors to address this issue in the literature, emphasizing the importance of preserving all elements of the anal canal or fistula tract to optimize postoperative continence and defecation (9). Levin proposed "the cutback procedure", which preserves all anal canal elements, reporting favorable bowel function outcomes in clinical follow-up (10).

This study showed that patients without trimming of the anorectum, and preservation of the dentate line structure did have significantly better functional outcomes. BFS was significantly lower, as mentioned in both bivariate as multivariate analyses. The lack of consensus in literature on terminology and surgical techniques, combined with the absence of studies on the impact of trimming on bowel function, underscores the need for further research.

Bowel function and Clinical Outcome

Almost half of the patients in this cohort achieved a normal bowel function (BFS of at least 18), with significant correlation between the perceptions of parents and the treating surgeon. In this study, patients with a rectoperineal fistula had better outcomes (median BFS: 17 [3; 20]) compared to those with a rectovestibular fistula (median BFS: 15 [4; 19]). However, a statistically significant difference was not observed, possibly due to the small sample size in the latter group. Additionally, as both rectovestibular and rectoperineal fistulas are low forms of ARM, significant differences between these two patient groups were not expected.

Beattie et al. conducted a comparable single-center study and reported that, in contrast to presented findings, more than half of their population had poor bowel function (22). Their study included both high and low forms of ARM, with significantly worse scores regarding both incontinence and constipation, however mainly present in the high ARM group. Peña et al. reported that when bowel management was appropriately applied 90% of fecal incontinence could be overcome, even in the less favorable ARM types (23). When comparing with a recent French multicenter study by Schmitt et al., including over 350 patients post-ARM repair, constipation rate was similar (41% versus 34% in this cohort) (24). Additionally, they reported the highest incidence of constipation in the group of 12-16 years old, with almost half of the adolescents affected. In this study, bowel function was not evaluated across different age groups, however literature states that constipation improves with age, potentially due to growth and hormonal changes during puberty (18, 25).

Regarding soiling, Schmitt et al. showed a higher prevalence, with 30-35% of patients experiencing occasional soiling once a week (versus 23% in this cohort), without significant age group differences (24). They did note a lower percentage of children experiencing social problems due to soiling, with a total of 6.5%, compared to 15% in this cohort.

Developmental disorder and associated anomalies

An important influencing factor on bowel function was the presence of a developmental disorder, with clearly lower BFS obtained. Pediatricians, surgeons and general practitioners should be aware when developmental disorders are present in ARM, to actively screen and manage underlying problems regarding bowel function. Incidence of associated anomalies are reported very differently in the literature. A recent Australian study by Evan-Barns et al. reported a higher number of associated anomalies (79%) and of VACTERL association (53%) (versus respectively 40% and 12.5% in this study). However, they showed that low forms of ARMs do not necessarily correlate with a lower incidence of other anomalies, and that these children are at risk for higher morbidity, as they are less likely to receive complete screening for associated anomalies (3).

Limitations

In this cohort PSARP was carried out in all patients before 2015. Although we obtained a large cohort sample from a single center, the subgroup analyses may have lacked sufficient statistical power due to the small group sizes. To asses bowel function, the Rintala questionnaire (12) was used, which is easy to use and interpret, but evaluating and comparing bowel function remains challenging due to the lack of standardized tools. Also, data were collected retrospectively, with assessments by parents and treating surgeons at different time-points, making comparisons challenging. Interpretation of parental reports may yield bias due to overestimation of good bowel function, as data were directly linked to the treating physician. Importantly, this study focused solely on bowel function and gastrointestinal outcomes, but urogenital function, sexual and reproductive health, mental health, and social acceptance should not be overlooked and need to be evaluated in the future.

Future perspectives

Our study suggests that avoiding anorectal trimming is preferable for better functional outcomes. However, studies on this subject are absent and further research with larger sample sizes is needed to confirm these findings.

Conclusion

This study focused on a homogenous group of low ARM cases treated with PSARP, showing that almost 50% achieved normal bowel function scores, with no gastrointestinal issues at a median follow-up age of 7.7 years. A worse bowel function seems to be present after anorectal trimming, suggesting it to be avoided during surgery when feasible. The timing of surgery remains debated, with a preference for early repair. The presence of a developmental disorder showed significant impairment of bowel function, and should be taken into consideration in follow-up and counseling. The Rintala questionnaire is useful at follow-up, additional to the clinical outcome evaluated by the treating surgeon. Long-term follow-up strategies should be adapted to monitor quality of life and adequately counsel patients and their parents.

The authors declare that they have no conflicts of interest.

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