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Determinants of Long-Term Bowel Function and Quality of Life Following PSARP for Anorectal Malformations

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Abstract

Purpose

Anorectal malformations (ARM) are associated with substantial variability in long-term bowel function. Although anatomical severity and associated anomalies are identified determinants of outcome, the influence of surgical timing remains controversial. This study evaluated predictors of long-term bowel function and health-related quality of life following posterior sagittal anorectoplasty (PSARP).

Methods

A national single-center cross-sectional study included patients with ARM treated by PSARP between 1997 and 2019 and aged ≥ 3 years at follow-up. Functional outcomes were assessed using the Bowel Function Score (BFS), and health-related quality of life using the Pediatric Quality of Life Inventory (PedsQL). Associations between outcomes, surgical timing, ARM subtype, and associated anomalies were assessed using uni- and multivariable analyses.

Results

Fifty-six patients were included (57% female). Median BFS was 16.0 (IQR 12.8–19.0); 39% achieved normal bowel function (BFS ≥ 18). Constipation and soiling were present in 46% and 57% of patients, respectively. Bowel function differed significantly among ARM subtypes (Kruskal-Wallis $H = 17.07$, $p = 0.004$). In multivariable analysis, spinal anomalies were independently associated with lower BFS (β −5.38, $p = 0.009$), whereas age at definitive reconstruction was not associated with bowel function ($p = 0.286$). Better bowel function correlated with higher PedsQL scores ($\rho = 0.43$, $p = 0.002$).

Conclusion

Long-term bowel function after PSARP is primarily determined by anatomical and neurological factors rather than surgical timing. Better bowel function is associated with improved quality of life, supporting long-term multidisciplinary follow-up in patients with ARM.

Introduction

Anorectal malformations (ARM) comprise a wide spectrum of congenital anomalies, affecting approximately 1 in 4000–5000 live births (1). Although the etiology of ARM is multifactorial and not yet fully understood (2), surgical reconstruction has dramatically improved survival and functional outcomes. Posterior sagittal anorectoplasty (PSARP) remains one of the standard surgical approaches (3), while laparoscopically assisted anorectal pull-through and sphincter-sparing techniques are increasingly applied in selected cases (4–6).

Despite technical progress, long-term bowel function (BF) outcomes remain heterogeneous. Functional impairment is influenced by anatomical complexity, development of the pelvic floor and sphincter complex, and the presence of associated anomalies, particularly spinal and sacral malformations (7–10). Some authors have suggested that earlier reconstruction might support improved neuromuscular development and functional outcomes (11); however, evidence remains inconsistent.

Patient-reported quality of life (PQoL) has emerged as a key outcome measure in patients with ARM. While bowel dysfunction is expected to negatively impact PQoL, multiple studies suggest that patients may adapt over time and report preserved quality of life despite ongoing functional limitations (12–17). Understanding the interplay between anatomical subtype, BF, and perceived quality of life is crucial to guide long-term management and counseling.

The primary aim of this study was to identify predictors of long-term BF following PSARP in patients with ARM. Secondary objectives were to evaluate the relationship between BF and PQoL and to assess whether timing of definitive reconstruction independently influences long-term outcomes.

Methods

A national, single center cross-sectional study was conducted. Inclusion criteria were reconstruction by PSARP, age ≥ 3 years at evaluation. Exclusion criteria included death, permanent stoma, severe neurological or cognitive impairment, or loss to follow-up.

BF was assessed using the Bowel Function Score (BFS) (10), and constipation and soiling were recorded. PQoL was evaluated using the Pediatric Quality of Life Inventory (PedsQL™) (18). Patient-reported total PedsQL scores (range 0–100) were used for primary analysis; parent-reported data were used as supportive information.

Predictors included ARM type (8), associated anomalies (VACTERL association, spinal anomalies, sacral anomalies), timing of definitive reconstruction, and timing of stoma closure.

Continuous variables were summarized as medians and ranges. Group comparisons were performed using the Mann-Whitney U test or Kruskal-Wallis's test. Correlations between BFS and PedsQL were assessed using Spearman's rank correlation coefficient. Uni and multivariable linear regression models were applied to identify independent predictors of BFS. Variables considered clinically relevant were included in the final model regardless of univariable significance.

The study was approved by the national ethics committee (Ethics approval: IRB No. 0120 – 14/2022/3).

Results

Fifty-six patients with ARM were included in the study; the main demographic and clinical characteristics are described (Table 1). Outcomes according to ARM subtype are described in Table 2. Anatomical

subtype significantly influences long-term BF outcomes (p = 0.004), but PQoL remained preserved and did not differ significantly across anatomical groups (p = 0.128).

Table 1

Demographic and clinical characteristics; BFS – Bowel function score, PedsQL - Pediatric Quality of Life Inventory. M – median, IQR – interquartile range.

Characteristic	Value
Number of patients	56
Female sex	32 (57%)
Age at definitive reconstruction M (IQR)	79 (6.8-182.3)
BFS M (IQR)	16 (12.8–19.0)
BFS ≥ 18	22 (39%)
Constipation	25 (46%)
Soiling	32 (57%)
Spinal anomalies	7 (14%)
Sacral anomalies	31 (55%)
Patient-reported PedsQL total score M (IQR)	84.1 (73.4–95.0)

Table 2

Outcomes according to ARM subtype; RPF – rectoperineal fistula, RVF – rectovestibular fistula, RBF – rectobulbar fistula, WF – without fistula, RpF – rectoprostatic fistula. BFS – Bowel function score, PedsQL - Pediatric Quality of Life Inventory score.

ARM subtype	n	Median BFS (IQR)	Median PedsQL (IQR)
RPF	23	19.0 (16.0–19.0)	85.3 (72.9–98.8)
RVF	15	14.0 (12.0-17.5)	76.9 (72.6–85.1)
RBF	9	11.0 (8.0–13.0)	85.3 (73.9–87.5)
WF	4	16.5 (12.3–19.0)	84.4 (84.1–92.2)
Cloaca	4	15.5 (15.0–17.0)	93.4 (90.4–96.3)
RpF	1	8.0	61.4
Kruskal-Wallis H for BFS = 17.07, p = 0.004			
Kruskal-Wallis H for PedsQL = 8.57, p = 0.128			

In univariable analyses, spinal anomalies (β − 5.61, p = 0.001) and sacral anomalies (β − 6.53, p = 0.006) were associated with significantly lower BFS scores. Older age at stoma closure was associated with

worse bowel function (β − 0.005, p = 0.038), whereas age at PSARP showed only borderline significance (p = 0.053) (Table 3). In multivariable analyses only spinal anomalies were associated with significantly worse BFS (β − 5.38, p = 0.009) (Table 4). As for predicting the PQoL, there was a significant positive correlation with BFS (β 1.35, p = 0,003), not the associated anomalies (Table 5).

Table 3
Univariable predictors of bowel function (BFS). PSARP – posterior sagital anorectoplasty.

Variable	β	95% CI	p
Spinal anomalies	-5.61	-8.82 to -2.41	0.001
Sacral anomalies	-6.53	-11.07 to -1.99	0.006
Age at PSARP (days)	-0.010	-0.021 to 0.000	0.053
Age at stoma closure (days)	-0.005	-0.010 to 0.000	0.038

Table 4
Multivariable predictors of bowel function. PSARP – posterior sagital anorectoplasty.

Variable	Adjusted β	95% CI	p
Age at PSARP	-0.005	-0.015 to 0.005	0.286
Spinal anomalies	-5.38	-9.35 to -1.42	0.009
Sacral anomalies	-2.99	-7.84 to 1.86	0.221
Model R^2 = 0.29			

Table 5
Predictors of health-related quality of life (PedsQL). BFS – bowel function score.

Variable	Adjusted β	95% CI	p
BFS	1.35	0.48 to 2.21	0.003
Spinal anomalies	-6.70	-18.39 to 4.99	0.255
Sacral anomalies	11.46	-5.81 to 28.72	0.189

Model R^2 = 0.24.

The relationship between BFS and PQoL is rather diverse, but shows positive correlation (Spearman's ρ = 0.43, p = 0.0015) (Fig. 1).

Scatterplot showing the relationship between Bowel Function Score (BFS) and patient-reported Pediatric Quality of Life Inventory (PQoL) total score in patients with anorectal malformations following posterior

sagittal anorectoplasty (PSARP). The solid line represents the fitted linear regression model. Higher BFS values were associated with better health-related quality-of-life scores *(Spearman's ρ = 0.43, p = 0.0015)*.

Discussion

Principal findings

The present study evaluated long-term BF and PQoL following PSARP in a national cohort of patients with ARM. Three principal findings emerged. First, BF differed significantly according to ARM subtype, confirming the importance of anatomical complexity in determining long-term outcomes. Second, spinal anomalies were independently associated with worse BF, whereas timing of definitive reconstruction was not an independent predictor. Third, better BF was associated with better PQoL, although quality of life scores did not differ significantly between anatomical subgroups.

Anatomical factors as determinants of bowel function

Long-term BF following ARM repair remains highly variable, even when similar surgical techniques are used. In our cohort, BF differed significantly between ARM subtypes (Kruskal-Wallis p = 0.004), with the highest BFS values observed in patients with RPF and lower values in patients with RBF, RpF, and complex malformations. Furthermore, spinal anomalies were associated with an adjusted reduction of more than five points in BFS.

These findings are consistent with previous studies demonstrating that long-term bowel outcomes are strongly influenced by the underlying anatomy and integrity of the sphincter complex, sacrum, and spinal cord (7, 19, 20).

Our findings support the concept that long-term bowel control is biologically constrained and cannot be explained solely by technical aspects of surgery.

Does the timing of reconstruction matter?

The optimal timing of definitive reconstruction remains debated. Some authors have hypothesized that earlier repairs may facilitate neuromuscular development and improve functional outcomes. Most recently, Pelizzo et al. suggested that earlier reconstruction could be associated with improved bowel control and consequently quality of life outcomes (11).

In contrast, our study found no independent association between age at PSARP and long-term BF after adjustment for anatomical and neurological factors. The regression coefficient for age at surgery was small and statistically non-significant, suggesting that surgical timing contributes little to outcome variability once underlying anatomy is considered.

This observation aligns with reports showing substantial heterogeneity in BF despite standardized surgical pathways (16, 17). Since complex malformations are often reconstructed later than simpler anomalies, apparent associations between timing and outcome may partly reflect confounding anatomical severity.

From a clinical perspective, our findings suggest that surgeons should avoid overemphasizing surgical timing as a dominant determinant of long-term BF. Careful preoperative assessment of spinal and sacral anatomy may be more informative for counselling families about expected outcomes.

BF and PQoL

A central strength of the present study is the simultaneous evaluation of functional outcomes and patient-reported quality of life. We observed a moderate positive correlation between BFS and PedsQL scores (Spearman $\rho = 0.43$, $p = 0.0015$), indicating that better BF is associated with improved PQoL.

This finding is consistent with previous studies. Authors reported that BF represents the major determinant of a patient's well-being. Patients experiencing constipation or soiling report lower psychosocial functioning and greater social limitations (12–14).

Interestingly, despite significant differences in BF among ARM subtypes, we found no statistically significant differences in overall quality of life scores between anatomical groups (Kruskal-Wallis $p = 0.128$). Previous studies have shown that many patients develop effective coping strategies and maintain satisfactory psychosocial functioning despite persistent bowel dysfunction, particularly when supported by structured bowel-management programs and multidisciplinary follow-up (19, 20). Moreover, successful symptom control rather than complete continence may be the most important determinant of long-term quality of life (16). Consistent with our findings, largely preserved health-related quality of life despite ongoing functional impairment in a substantial proportion of patients with anorectal malformations was reported (14).

Our findings therefore reinforce the importance of incorporating patient-reported outcome measures (PROMs) into long-term follow-up protocols rather than relying exclusively on objective functional scores (21).

Clinical implications

The results of this study have several practical implications.

First, patients with spinal anomalies appear to represent a particularly high-risk subgroup and may benefit from earlier involvement of multidisciplinary bowel-management programs.

Second, the lack of an independent association between surgical timing and BF suggests that moderate variations in timing of reconstruction are unlikely to alter long-term functional outcomes.

Third, because quality of life remains preserved in many patients despite functional limitations, long-term management should focus not only on BF but also on psychosocial adaptation, self-esteem, participation in daily activities, and family support.

Strengths and limitations

The strengths of this study include the use of a national cohort, long-term follow-up, uniform treatment within a single tertiary referral center, and simultaneous assessment of both BF and PQoL using validated instruments.

Several limitations should also be acknowledged. The retrospective cross-sectional design limits causal inference. The sample size was small, reflecting the rarity and heterogeneity of ARM. Some analyses may have been underpowered, particularly within individual ARM subgroups. The study relied primarily on questionnaire-based assessment and did not include objective physiological measures such as anorectal manometry, colonic transit studies, or detailed radiological evaluation of pelvic floor anatomy. Finally, potential patient response bias cannot be excluded.

Overall, our findings support the view that long-term BF after PSARP is determined by anatomical and neurological factors rather than timing of reconstruction. Although bowel dysfunction remains common, PQoL is preserved and linked to functional bowel outcomes. Future multicenter studies with larger cohorts and standardized outcome assessment are needed to further define prognostic factors and optimize long-term management strategies for patients with anorectal malformations.

Conclusion

Long-term outcomes after PSARP appear to be driven more by anatomy than by timing of reconstruction. While spinal and anatomical abnormalities significantly influence bowel function, quality of life remains preserved and is more linked to functional status than to anatomical subtype. These findings highlight the need for early identification of high-risk patients and sustained multidisciplinary follow-up extending beyond childhood.

Declarations

Ethics Approval

This study was conducted in accordance with the principles of the Declaration of Helsinki and was approved by the National Medical Ethics Committee of Slovenia (IRB No. 0120-14/2022/3). Written informed consent was obtained from all participants and/or their legal guardians, where applicable.

Author Contributions

Polona Studen Pauletić: Conceptualization, methodology, data collection, formal analysis, interpretation of data, manuscript drafting, and manuscript revision.

Aleš Tomažič: Methodology, interpretation of data, critical revision of the manuscript, and supervision.

Matjaž Homan: Study design, interpretation of data, critical revision of the manuscript, and supervision.

All authors reviewed and approved the final version of the manuscript.

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Conflict of Interest

The authors declare that they have no conflict of interest.

Data Availability Statement

The datasets generated and/or analysed during the current study are not publicly available because they contain personal health information and are subject to institutional and national data protection regulations. De-identified data may be available from the corresponding author upon reasonable request and with permission of the University Medical Centre Ljubljana.

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Use of Artificial Intelligence

The authors used artificial intelligence-assisted language tools exclusively for language editing and manuscript preparation. All analyses, interpretations, conclusions, and final manuscript content were reviewed and approved by the authors, who take full responsibility for the submitted work.

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Figures

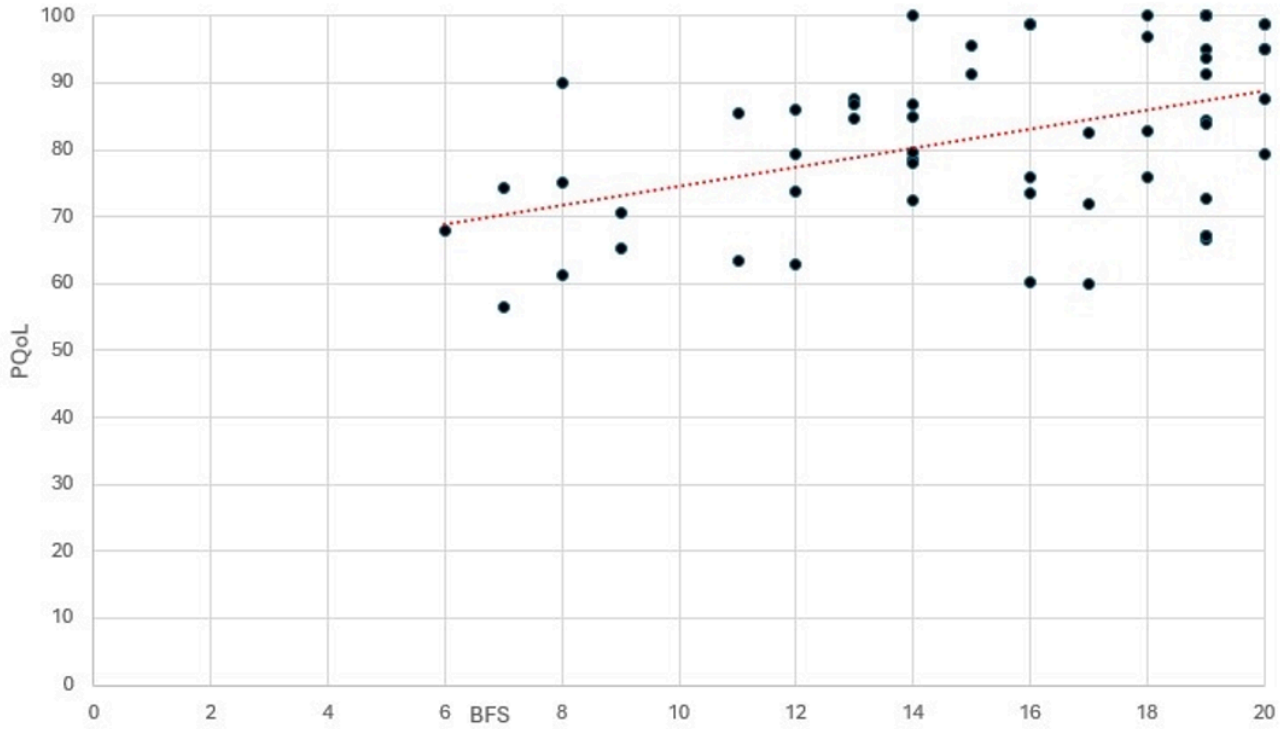


Figure 1

Association between bowel function and patient-reported quality of life following PSARP.