



Gastrointestinal quality of life and bowel function in adults born with anorectal malformation and hirschsprung disease

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Abstract

Purpose To assess the quality of life and disease-specific functioning of adults with anorectal malformations (ARM) or Hirschsprung disease (HD) compared to healthy reference scores.

Methods Patients with the diagnosis of ARM or HD from the Adult Colorectal Research Registry completed the Short Form 36 Health Survey (SF-36), the Gastrointestinal Quality of Life Index (GIQoLI), and the Bowel Function Score (BFS) between October 2019 and August 2022. One-sample Wilcoxon test compared the results to reported healthy references with a significance level of < 0.05 .

Results The response rate was 67%. All three surveys were completed by 133 adults with a slight preponderance of males (51%). Median age was 31 years, 117 were born with ARM and 16 with HD. All subgroups had significantly lower BFS than healthy references. ARM patients scored significantly lower than the healthy reference population when assessed for GIQoLI. All showed significant impairment with the mental component summary (MCS) of SF-36. Patients with a successful bowel management had significantly higher scores on all three questionnaires than those with fecal accidents.

Conclusion Our results emphasize the importance of a successful bowel management and its impact on the quality of life and bowel function. Long-term follow-up is recommended with attention to mental health.

Keywords Anorectal malformation · Imperforate anus · Hirschsprung disease · Quality of life · Long-term follow-up

Introduction

Patients born with an anorectal malformation (ARM) or Hirschsprung disease (HD) require surgical treatment in childhood. In patients with ARM, fecal incontinence may occur despite correct surgical repair, as this also depends on the severity and type of the ARM and associated anomalies [1]. In patients with HD fecal control should not be impaired after surgery provided the anal canal is intact [2, 3].

Quality of life and bowel function in patients born with ARM or HD has been previously reported for children by their parents or caregivers and by adolescent and adult patients [4–15]. Long-term studies of adult patients with ARM have shown that fecal continence is a strong predictor of quality of life (QoL) [11]. For those with HD, bowel function has been shown to deteriorate with age, with only slight impact on QoL [12]. Conversely, it has been observed that adult patients with ARM reported an improvement in fecal incontinence or soiling over time [9]. It remains unclear if this improvement is real, or if it is due to an inherent adaptation to the individual's condition and change in their expectations [8, 11]. However, it has also been shown that soiling, constipation, associated anomalies, severity of ARM, and presence of abdominal pain, all correlated significantly with patients' psychosocial morbidity [16]. Treatment of fecal incontinence with a comprehensive bowel management program was described in 2009 by the senior authors [17]. Its impact on the quality of life in children with fecal incontinence has previously been reported [18]. Three months

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after implementation of a successful bowel management program, the fecal incontinence index (FII) as well as the quality of life (PedsQL 4.0) have shown statistically significant improvement.

Given the importance of fecal control for patient wellbeing, this study sought to evaluate gastrointestinal quality of life, bowel function and the influence of successful bowel management in adult patients with ARM and HD. Three validated instruments were used, Bowel Function Score (BFS), 36-item short-form health survey (SF-36) and Gastrointestinal Quality of Life (GIQoL), which assesses bowel function, physical and mental health components and quality of life system specific in patients with gastrointestinal disorders. It was hypothesized that adult patients with ARM or HD that experienced fecal incontinence without proper bowel management would report lower levels of gastrointestinal quality of life compared to a healthy population.

Methods

This was a cross sectional study of male and female patients born with either an ARM or HD who underwent surgical intervention by our team. Patients who were older than 18 years of age in the Adult Colorectal Research Registry were contacted to participate in the research study to assess bowel function and quality of life. Patients were sent electronic surveys through REDCap. REDCap is a secure, web-based application to capture data for clinical research [19]. Study enrollment occurred between October 2019 and August 2022. In addition to basic demographic information, general health, bowel management, fecal control, stool accidents and soiling were assessed. Patients were asked to complete three validated questionnaires: Bowel Function Score (BFS), the Gastrointestinal Quality of Life Index (GIQoL) and the Short Form 36 Health Survey (SF-36).

Bowel function score (BFS)

Bowel function score (BFS) is a 7-item multivariate scoring system, including questions of fecal control and social impact due to bowel habits [5]. The participants score each question from zero to three, except for the question of stool frequency, which is scored from one to two. The scores are then summarized with a possible maximum score of 20, indicating good bowel control.

Gastrointestinal quality of life index (GIQOLI)

Gastrointestinal quality of life index (GIQOLI) was validated in 1995 and assesses the four dimensions of gastrointestinal symptoms, physical and social function, and emotional status. The participants answer each question on a five-point Likert scale from zero to four. Values are summed with a maximum score of 144 [20].

Short form 36 health survey (SF-36)

Short form 36 health survey (SF-36) was developed in 1992 and assesses eight health concepts, which are physical functioning, role limitations due to physical or emotional health problems, bodily pain, general health, vitality, social functioning, and mental health [20]. These scores are then presented as a physical component summary (PCS) and a mental component summary (MCS) subscore and understood as the physical and mental components of health, although these should always be interpreted in the context of all subscales [21, 22].

Patients were included in the study if they were confirmed to have a diagnosis of ARM or HD and completed all three questionnaires. The reported demographic data of the patients was linked to their surgical records to ensure accurate colorectal diagnoses. Bowel function and control was assessed and defined as clean, if no involuntary bowel movements occurred (fecal accidents or soiling), and not clean, when the patient reported to have fecal accidents or soiling, despite having a bowel regimen of either laxatives or enemas (antegrade or retrograde). Patients with an ostomy were excluded from the analysis of the BFS and analyzed separately.

Survey scores were summarized and reported as median (Q1, Q3) and compared to reported results of a healthy comparison study population in literature [20, 23, 24]. In addition, analysis was also performed comparing outcomes between the study population groups of ARM and HD to measures of patients with inflammatory bowel disease (IBD) reported in the literature [25–27].

One-sample Wilcoxon tests were used to compare BFS, GIQoL, and SF-36 scores to reference values from healthy and IBD populations [20, 23–28]. In some cases, the healthy reference values were the maximum possible subscore and thus no formal testing was conducting. Statistically significant p-values (<0.05) indicate that the median score for a

given diagnosis group within our study sample is significantly different from the reference value.

Additionally, two-sample Wilcoxon tests or Kruskal–Wallis tests were used to compare scores between bowel control subgroups. For statistically significant (p -value < 0.05) Kruskal–Wallis tests, post-hoc Dunn’s tests were used to identify statistically significant differences in pairwise comparisons between bowel control subgroups. Due to the exploratory nature of this study, p -values were not adjusted for multiple comparisons. Statistical analyses were performed using R Studio (versions 4.1.2). This study was approved by the Colorado Multiple Institutional Review Board (COMIRB #19–1050, 19–0899, 21–3153, 21–3154).

Results

Demographic summary

A total of 220 patients were contacted to participate in the study and 147 responded (67% response rate). There were 14 patients excluded due to missing documentation, diagnosis other than ARM or HD or because they did not answer all three questionnaires. One hundred and thirty-three adults completed all three surveys, with 63 females (47.4%), 68 males (51.1%), and two nonbinary participants (1.5%) (Table 1). Age ranged from 22 to 80 years, with a median of 31 years and a mean of 32.5 years. One hundred and seventeen were born with an ARM, of those, 38 (32.5%)

were cloaca and seven (6%) had a complex malformation. Sixteen (12%) were born with HD. Four patients (3%) had a kidney transplant, three of whom were born with a cloaca. Thirty-six patients (27.1%) had an antegrade continence enema (ACE) procedure and seven patients (5.3%) have a permanent ostomy (colostomy or ileostomy).

Of these seven patients with an ostomy, six were female, one was male and their diagnoses included cloaca (4), covered cloacal exstrophy (1), total colonic aganglionosis (1), and ARM with recto-urethral prostatic fistula (1).

Bowel function score (BFS—Table 2)

All subgroups had significantly lower composite scores when compared to healthy reference values. Within the subscores statistically significant impairment was found for patients with cloaca and ARM for all categories except for stool frequency and social problems.

Gastrointestinal quality of life (GIQoL—Tables 3, 4, 5)

Patients with ARM, cloaca or complex malformations had significantly lower composite scores when compared to healthy reference values. Patients with the diagnosis of cloaca (median score 99) or complex malformations (median score 100) had even lower composite scores than patients suffering from Crohn’s

Table 1 Demographic summary

	ARM (N = 72)	Cloaca (N = 38)	Complex malformation (N = 7)	Hirschsprung disease (N = 16)	Overall (N = 133)
Gender					
Female	17 (23.6%)	36 (94.7%)	6 (85.7%)	4 (25.0%)	63 (47.4%)
Male	55 (76.4%)	0	1 (14.3%)	12 (75.0%)	68 (51.1%)
Non-binary	0	2 (5.26%)	0	0	2 (1.50%)
Age					
Median (Q1, Q3)	30.0 (25.0, 37.0)	31.0 (27.0, 35.0)	35.0 (26.0, 39.0)	29.5 (25.8, 32.3)	31.0 (26.0, 36.0)
Kidney transplant					
No	71 (98.6%)	35 (92.1%)	7 (100%)	16 (100%)	129 (97.0%)
Yes	1 (1.39%)	3 (7.89%)	0	0	4 (3.01%)
Antegrade procedure					
No	56 (77.8%)	25 (65.8%)	5 (71.4%)	11 (68.8%)	97 (72.9%)
Yes	16 (22.2%)	13 (34.2%)	2 (28.6%)	5 (31.3%)	36 (27.1%)
Ostomy					
No	71 (98.6%)	34 (89.5%)	6 (85.7%)	15 (93.8%)	126 (94.7%)
Yes	1 (1.39%)	4 (10.5%)	1 (14.3%)	1 (6.25%)	7 (5.26%)

ARM Anorectal malformation

Table 2 Bowel function score (BFS)

Score	Diagnosis	Median (Q1, Q3)	Healthy reference	
			Reference value	P-value
Composite score	ARM	14 (12, 17)	19.2	<0.001
	Cloaca	12 (9, 16)		<0.001
	Complex malformation	15.5 (13.5, 16)		0.031
	Hirschsprung disease	15 (12.5, 18)		0.001
Hold back defecation	ARM	2 (1, 3)	2.94	<0.001
	Cloaca	2 (0.25, 2)		<0.001
	Complex malformation	2 (2, 2)		0.062
	Hirschsprung disease	2 (2, 3)		0.075
Feels urge to defecate	ARM	2 (1, 2)	2.91	<0.001
	Cloaca	2 (1, 2)		<0.001
	Complex malformation	3 (2.25, 3)		0.812
	Hirschsprung disease	2 (2, 3)		0.027
Stool frequency	ARM	2 (1, 2)	1.91	0.541
	Cloaca	2 (1, 2)		0.118
	Complex malformation	2 (2, 2)		0.531
	Hirschsprung disease	2 (1, 2)		0.782
Soiling	ARM	2 (1, 3)	2.63	<0.001
	Cloaca	2 (1, 2)		<0.001
	Complex malformation	2 (1.25, 2.75)		0.125
	Hirschsprung disease	2 (2, 3)		0.005
Accidents	ARM	2 (2, 3)	2.94	<0.001
	Cloaca	2 (2, 3)		<0.001
	Complex malformation	2 (2, 2.75)		0.125
	Hirschsprung disease	2 (2, 3)		0.07
Constipation	ARM	2 (1, 3)	2.89	<0.001
	Cloaca	2 (1, 3)		<0.001
	Complex malformation	2 (2, 2.75)		0.125
	Hirschsprung disease	3 (2, 3)		0.183
Social problems	ARM	3 (2, 3)	2.94	0.15
	Cloaca	2.5 (1, 3)		0.011
	Complex malformation	3 (2.25, 3)		1
	Hirschsprung disease	2 (1.5, 3)		0.066

One-sample Wilcoxon test results

Kyrklund K, Pakarinen MP, Rintala RJ (2017) Long-term bowel function, quality of life and sexual function in patients with anorectal malformations treated during the PSARP era. *Semin Pediatr Surg*; 26: 336–342, <https://doi.org/10.1053/j.sempedsurg.2017.09.010>

Table 3 GIQoL composite score

Diagnosis	Median (Q1, Q3)	Reference value healthy[20]	p-value
ARM	120 (103.25, 130.25)	125.8	<0.001
Cloaca	99 (81.25, 119)	125.8	<0.001
Complex malformation	100 (89, 110.5)	125.8	0.031
Hirschsprung	125 (94, 131.25)	125.8	0.226

One-sample Wilcoxon test results for comparison of patients with healthy reference values

disease (reference score 103), or ulcerative colitis (reference score 110) [20, 25, 26]. In contrast, the HD group reached similar composite scores (reference score 125) as the healthy population (reference score 125.8).

SF-36 (Table 6)

Patients with cloaca were the only subgroup who had statistically significant lower scores for PCS. All patients scored lower for MCS, compared to healthy reference values, indicating impaired mental health. Patients with

Table 4 GIQoL composite score

Diagnosis	Median (Q1, Q3)	Reference value of IBD/CD [25]	p-value
ARM	120 (103.25, 130.25)	103.0	<0.001
Cloaca	99 (81.25, 119)	103.0	0.296
Complex malformation	100 (89, 110.5)	103.0	0.688
Hirschsprung disease	125 (94, 131.25)	103.0	0.188

One-sample Wilcoxon test results for comparison of patients with Crohn's Disease versus study population

IBD inflammatory bowel disease, CD Crohn's disease

Table 5 GIQoL composite score

Diagnosis	Median (Q1, Q3)	Reference value of IBD/UC[26]	p-value
ARM	120 (103.25, 130.25)	110.0	0.004
Cloaca	99 (81.25, 119)	110.0	0.007
Complex malformation	100 (89, 110.5)	110.0	0.297
Hirschsprung disease	125 (94, 131.25)	110.0	0.733

One-sample Wilcoxon test results for comparison of patients with Ulcerative Colitis versus study population

ARM anorectal malformation, IBD inflammatory bowel disease, UC ulcerative colitis

complex malformations, had the lowest scores for PCS and MCS within the subgroups. In comparison to patients with IBD, patients with complex malformations had lower scores across subscales, except for the subscores of role functioning-physical and emotional wellbeing.

Outcome based on fecal control (Tables 7, 8, 9, 10)

The study population was stratified by functional outcome (fecal accidents and soiling) and bowel management (either laxatives or enemas) regardless of their diagnosis. The majority of patients (51%) were clean with or without laxatives and 9.8% were clean with enemas (Table 7). Fecal accidents with laxative treatment occurred in 7.5% of patients and in 11.3% with enemas. Twenty patients who were having fecal accidents did not undergo formal bowel management (neither laxatives nor enemas) were excluded from the analysis.

Patients with bowel control or successful bowel management with laxatives had the highest scores among the patient subgroups, followed by patients who were clean on daily

enemas (Table 8). There was a statistically significant difference the scores of BFS, GIQoL and PCS among the subgroups. This did not apply to MCS where all subgroups had similar low scores regardless of the bowel management type and outcome. Patients with an unsuccessful bowel management either on laxatives or enemas scored even lower than patients with an ostomy in the MCS.

When the patients were grouped as clean and not clean in the underwear (Table 9), the patients who stayed clean had statistically significantly higher scores ($p < 0.05$) than those who had fecal accidents, on all three questionnaires. Compared to healthy reference scores for all three validated instruments (BFS, SF-36 and GIQoL) all patients, regardless if they were clean or not clean in the underwear, scored significantly lower throughout all questionnaires (Table 10).

Ostomy

Seven patients had an ostomy. One respondent stated that she had the ostomy by choice and, but no further information was provided by the other six participants about why they had an ostomy. Significant differences in GIQoL and PCS score were seen when patients with an ostomy were compared to patients who were clean in the underwear (with or without laxatives). Ostomy patients had significantly lower scores (unadjusted p-values < 0.05) (Table 11). There was no statistical significance in GIQoL and PCS in patients with unsuccessful bowel management and accidents compared to patients with an ostomy (Table 11).

Discussion

This study assessed bowel function, gastrointestinal quality of life, and general health of adult ARM and HD patients using three different validated questionnaires. When available, scores were also compared with patients with inflammatory bowel disease, since this population also suffers from a chronic condition that impacts the gastrointestinal tract.

Consistent with prior published data [7], all patients in this study reported significant impairment on the BFS, suggesting poor bowel functioning and control. It is known that patients with ARM and HD require long-term follow-up, because even when they receive a technically correct operation, 75% of patients will require bowel management, most commonly laxatives or enemas. Due to the nature of this study, many enrolled patients were lost to follow-up at the time that questionnaires were sent and were not offered, or declined, bowel management. We believe that the next generation of adult patients will have stricter follow-up, as well as a more structured bowel management regimen, thanks to the spread of knowledge, awareness

Table 6 Short form-36

SF-36			Healthy reference [22]		IBD reference [27]	
Score	Diagnosis	Median (Q1, Q3)	Reference value	P-value	Reference value	P-value
PCS	ARM	55.4 (51.0, 57.9)	55.85	0.062	n/a	n/a
	Cloaca	49.1 (39.4, 52.8)		<0.001		
	Complex malformation	47.3 (32.0, 52.5)		0.078		
	Hirschsprung disease	56.4 (53.4, 58.8)		0.744		
MCS	ARM	50.3 (38.7, 55.2)	54.74	<0.001	n/a	n/a
	Cloaca	44.5 (31.3, 53.0)		<0.001		
	Complex malformation	38.5 (33.3, 50.5)		0.016		
	Hirschsprung disease	50.2 (37.8, 55.4)		0.029		
Physical functioning	ARM	100 (90, 100)	90	<0.001	90	<0.001
	Cloaca	85 (65, 98.8)		0.035		0.035
	Complex malformation	85 (65, 87.5)		0.156		0.156
	Hirschsprung disease	100 (95, 100)		0.006		0.006
Role functioning- physical	ARM	100 (100,100)	100	n/a	25	<0.001
	Cloaca	75 (25, 100)				<0.001
	Complex malformation	75 (12.5, 100)				0.156
	Hirschsprung disease	100 (100, 100)				<0.001
Role functioning- emotional	ARM	100 (66.7, 100)	100	n/a	62	<0.001
	Cloaca	66.7 (33.3, 100)				0.348
	Complex malformation	33.3 (33.3, 100)				0.859
	Hirschsprung disease	100 (75, 100)				0.577
Energy fatigue	ARM	60 (45, 75)	65	0.009	62.5	0.194
	Cloaca	45 (25, 60)		<0.001		<0.001
	Complex malformation	40 (40, 62.5)		0.094		0.109
	Hirschsprung disease	57.5 (43.8, 71.3)		0.14		0.280
Emotional well-being	ARM	74 (56, 84)	80	<0.001	50	<0.001
	Cloaca	66 (44, 84)		<0.001		0.005
	Complex malformation	56 (48, 78)		0.062		0.281
	Hirschsprung disease	76 (58, 88)		0.177		0.001
Social functioning	ARM	93.8 (71.9, 100)	100	n/a	66.7	<0.001
	Cloaca	75 (50.0, 87.5)				0.653
	Complex malformation	50 (43.8, 75)				0.156
	Hirschsprung disease	87.5 (75, 100)				0.126
Bodily pain	ARM	90 (69.4, 100)	74	<0.001	74	<0.001
	Cloaca	73.8 (59.4, 90)		0.782		0.782
	Complex malformation	45 (22.5, 83.8)		0.141		0.141
	Hirschsprung disease	90 (90, 100)		0.238		0.238
General health	ARM	72.5 (55, 80)	72	0.296	60	<0.001
	Cloaca	50 (25, 58.8)		<0.001		<0.001
	Complex malformation	45 (30, 72.5)		0.141		0.453
	Hirschsprung disease	80 (48.8, 86.3)		0.890		0.087

One-sample Wilcoxon test results for comparison with healthy and IBD-reference values

n/a Due to the distribution of the sample size, statistical significance was not assessed

ARM anorectal malformation, PCS physical component score, MCS mental component score, IBD inflammatory bowel disease

of patients and families, availability of colorectal centers offering bowel management, and initiatives for proper transition of care.

Regarding quality of life, patients with cloaca and other types of anorectal malformation had statistically significant lower scores compared to the healthy reference population.

Table 7 Fecal accidents and soiling based on bowel management stratified by diagnosis

	Clean with or without laxatives (N = 68)	Not Clean with laxatives (N = 10)	Clean with enemas (N = 13)	Not clean with enemas (N = 15)	Ostomy (N = 7)	Not clean, no BM (N = 20)
Diagnosis						
ARM	38 (55.9%)	7 (70%)	7 (53.8%)	6 (40%)	1 (14.3%)	13 (65%)
Cloaca	16 (23.5%)	3 (30%)	3 (23.1%)	8 (53.3%)	4 (57.1%)	4 (20%)
Complex malformation	3 (4.4%)	0	1 (7.7%)	0	1 (14.3%)	2 (10%)
Hirschsprung disease	11 (16.2%)	0	2 (15.4%)	1 (6.7%)	1 (14.3%)	1 (5%)
Laxatives						
No	57 (83.8%)	0	12 (92.3%)	14 (93.3%)	6 (85.7%)	20 (100%)
Yes	11 (16.2%)	10 (100%)	1 (7.7%)	1 (6.7%)	1 (14.3%)	0
Enemas						
No	68 (100%)	10 (100%)	0	0	5 (71.4%)	20 (100%)
Yes	0	0	13 (100%)	15 (100%)	2 (28.6%)	0
Bowel management						
No	51 (75%)	0	0	0	4 (57.1%)	17 (85%)
Yes	17 (25%)	10 (100%)	13 (100%)	15 (100%)	3 (42.9%)	3 (15%)

ARM anorectal malformation, BM bowel management

Table 8 Comparison of survey scores by bowel control subgroup, with Kruskal–Wallis test p-values

	Clean with or without laxatives (N = 68)	Not clean with laxatives (N = 10)	Clean with enemas (N = 13)	Not clean with enemas (N = 15)	Ostomy* (N = 7)	P-value
BFS composite score						
Median (Q1, Q3)	16 (14, 18)	9 (7.25, 12)	13 (12, 17)	9 (7, 10.5)	na*	<0.001
GIQoL composite score						
Median (Q1, Q3)	124 (110, 132)	93 (83.5, 107)	121 (116, 128)	98.0 (87.5, 108)	95.0 (88.5, 109)	<0.001
SF-36 physical component summary score (PCS)						
Median (Q1, Q3)	55.5 (51.3, 57.9)	53.2 (47.7, 54.6)	52.0 (49.9, 58.5)	47.5 (37.1, 53.6)	47.4 (37.4, 49.8)	<0.001
SF-36 mental component summary score (MCS)						
Median (Q1, Q3)	51.4 (42.8, 55.8)	40.5 (29.9, 57)	46.5 (40.6, 51.6)	40.7 (35, 52.6)	44.5 (33.4, 49.1)	0.343

BFS bowel function score, GIQoL gastrointestinal quality of life, SF-36 short form-36, na not available

*Patients with ostomy were excluded from the analysis of BFS

Table 9 Comparison of survey scores for clean in underwear (clean with/without laxatives or clean with enemas) vs. not clean in underwear (not clean with laxatives, not clean with enemas, or osteotomies), with two-sample Wilcoxon test p-values

	Clean in underwear (N = 81)	Not clean in underwear (N = 45)	P-value (N = 0)
BFS: composite score			
Median (Q1, Q3)	16.0 (14.0, 17.0)	10.0 (8.00, 12.0)	<0.001
GIQoL: composite score			
Median (Q1, Q3)	124 (110, 131)	98.0 (82.0, 110)	<0.001
SF-36: physical component summary score			
Median (Q1, Q3)	55.4 (51.0, 57.9)	49.8 (41.2, 55.4)	<0.001
SF-36: mental component summary score			
Median (Q1, Q3)	51.2 (42.4, 55.3)	39.4 (31.4, 52.5)	0.008

BFS bowel function score, GIQoL gastrointestinal quality of life, SF-36 short form-36

Table 10 One-sample Wilcoxon tests comparing clean and not clean bowel control subgroups to healthy reference

Bowel control subgroup	Median (Q1, Q3)	Healthy reference value	p-value
BFS composite score			
Clean	16 (14, 17)	19.20	<0.001
Not Clean	9 (7, 12)	19.20	<0.001
GIQoL composite score			
Clean	124 (110, 131)	125.80	0.011
Not Clean	98 (85, 107)	125.80	<0.001
SF-36 physical component summary score (PCS)			
Clean	55.36 (50.97, 57.92)	55.85	0.034
Not clean	49.77 (38.49, 54.25)	55.85	<0.001
SF-36 mental component summary score (MCS)			
Clean	51.2 (42.35, 55.33)	54.74	<0.001
Not clean	40.65 (34, 55.62)	54.74	<0.001

BFS bowel function score, *GIQoL* gastrointestinal quality of life, *SF-36* short form-36

A possible explanation is again the fact that even in malformations with good prognosis for bowel control, who have received a technically correct operation, bowel management with daily routines is still needed. Previous reports of adult patients born with ARM and HD scored lower in GIQoL compared to a healthy population, indicating that bowel control affects quality of life [4, 8, 11, 12]. Our study did not confirm these findings for patients with HD, who in this study, had a similar score to the healthy population. We believe this may be due to the fact that HD is usually an isolated condition, and when the surgery is done correctly, these patients do not suffer from fecal incontinence, even though some of them may require laxatives.

As we chose patients with inflammatory bowel disease as a comparison group for GIQoL, and most scores had no statistical significance, we should also acknowledge that the majority of IBD cases have a later onset than ARM and HD (that are typically diagnosed at birth). This difference may reflect upon the ability to cope and adapt for patients with ARM and HD.

When examining health concepts, ARM and HD patients reported the same level of physical impairment in their daily life as healthy references. On the other hand, all patients showed significant lower scores, concerning for notable impairment of mental health in this population. These findings are supported by a recent study that reported ARM patients had a higher prevalence of depression and anxiety than the general population [28]. No significant association was found in regards of type and severity of malformation with the prevalence of depression or anxiety. This is also supported by our data, that regardless

of the type of diagnosis, patients with congenital ARM or HD experience mental health problems. These findings highlight the importance of a multidisciplinary clinic, with long-term follow up including psychosocial providers to support those individuals who face mental health challenges, as a result of their congenital colorectal diagnoses. In addition, patients with ARM, cloacal anomaly, and complex malformations, frequently have associated anomalies that may impact their social functioning. For example, the need for intermittent catheterization, and frequency of UTI, these would not be commonly present for patients with HD and IBD.

Besides comparing patients by their diagnosis, patients were additionally compared by their bowel function, success of bowel management and fecal control. In both subgroups, clean versus not clean in the underwear, scoring was significantly worse than healthy references; however, the population who stayed clean in the underwear had scores closer to the healthy reference values. These results highlight the importance of a successful bowel management in patients born with ARM or HD, which likely leads to better overall functioning and quality of life. On the other hand, they also show that patients with an ostomy had similar scores as patients with unsuccessful bowel management. While it is challenging to understand why patients with ostomies may have impaired quality of life, it may be that this patient population is more medically complex or that the presence of an ostomy can affect on the patient's functioning. Further studies are warranted to assess and compare patients with an ostomy versus patients with unsuccessful bowel management. One of the seven patients with an ostomy, stated that she had the ostomy by choice. Due to the small sample size, no statement can be made, but we could postulate that a patient who gets an ostomy by choice, might not have as low of scores in these assessed areas, compared to patients who required an ostomy.

Limitations of this study are the small sample size with respect to the specific types of ARMs. We were underpowered to compare them adequately, we suspect, the prognosis for bowel control and level of complexity will have a direct impact on functioning and quality of life. All results were self-reported data and therefore subject to various biases, however this was minimized by the use of validated instruments. In addition, this study was conducted during the COVID pandemic, where social restrictions occurred, which may have impacted the overall quality of life and mental health of the participants, to a various degree [29]. Since this is a cross-sectional study and reflects only a single evaluation in these adult patients, further longitudinal studies are warranted to better assess this patient population over time.

Table 11 Dunn's test results for pairwise comparisons of statistically significant Kruskal–Wallis results from Table 8

Subgroups being compared	Unadjusted p-value	Bonferroni-adjusted p-value
BFS composite score		
Clean with enemas vs. clean with or without laxatives	0.093	0.186
Clean with enemas vs. not clean with enemas	<0.001	0.004
Clean with or without laxatives vs. not clean with enemas	<0.001	<0.001
Clean with enemas vs. not clean with laxatives	0.006	0.017
Clean with or without laxatives vs. not clean with laxatives	<0.001	<0.001
Not clean with enemas vs. not clean with laxatives	0.829	0.829
GIQoL composite score		
Clean with enemas vs. clean with or without laxatives	0.798	1
Clean with enemas vs. not clean with enemas	0.016	0.112
Clean with or without laxatives vs. not clean with enemas	<0.001	0.005
Clean with enemas vs. not clean with laxatives	0.014	0.108
Clean with or without laxatives vs. not clean with laxatives	<0.001	0.009
Not clean with enemas vs. not clean with laxatives	0.759	1
Clean with enemas vs. ostomy	0.081	0.407
Clean with or without laxatives vs. ostomy	0.024	0.146
Not clean with enemas vs. ostomy	0.833	0.833
Not clean with laxatives vs. ostomy	0.653	1
SF-36 physical component summary score (PCS)		
Clean with enemas vs. clean with or without laxatives	0.767	0.767
Clean with enemas vs. not clean with enemas	0.027	0.188
Clean with or without laxatives vs. not clean with enemas	0.001	0.011
Clean with enemas vs. not clean with laxatives	0.181	0.907
Clean with or without Laxatives vs. not clean with laxatives	0.054	0.326
Not clean with enemas vs. not clean with laxatives	0.498	1
Clean with enemas vs. ostomy	0.027	0.213
Clean with or without laxatives vs. ostomy	0.004	0.04
Not clean with enemas vs. ostomy	0.662	1
Not clean with laxatives vs. ostomy	0.333	1

SF-36 MCS was not provided, since this was shown that there was no statistical significance within the subgroups

BFS bowel function score, *GIQoL* gastrointestinal quality of life, *SF-36* Short form-36

Conclusion

Patients with ARM and HD reported an impairment of quality of life measurements compared to healthy controls. This impairment is associated with poorer GI/fecal control symptoms which indicate that proper bowel management can positively impact overall quality of life. Our study also emphasizes the importance of long-term follow-up with special attention to mental health support.

Author contributions L.A.W. and M.L.R. wrote the main manuscript text. L.A.W., J.M.R. and E.H.C. prepared the tables. All authors reviewed the manuscript.

Data availability The datasets generated and analyzed during the current study are available from the corresponding author on reasonable request.

Declarations

Competing interests The authors declare no competing interests.

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