RESEARCH ARTICLE

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Assessment of Quality of Life in Patients with Anorectal Malformations

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Abstract

Objective: Our main purpose in this study is to examine the effects of patients on their quality of life (QOL) according to anorectal malformations (ARM) types, and to compare them to patients with stool incontinence in the healthy population.

Methods: The research group was selected from 116 patients who were operated due to ARM between 2004-2018 and fulfilled the inclusion criteria, in University of Health Science İzmir Dr. Behçet Uz Pediatric Diseases and Surgery Training and Research Hospital. The control group was composed of 12 participants among 348 children between the ages 5-15 who were operated for phimosis in 2018 -2019. Fecal Incontinence Related Life Quality Questionnaire (QQVCFCA) scales were applied to both groups. SPSS 22.0 program was used.

Results: The research group was completed with 75 participants and the control group with 12 participants. The QQVCFCA score of the low-type ARM was significantly higher than the participants with the high-type ARM (p <0.001). The QQVCFCA score was lower in patients with additional anomalies, poor physical examination, presence of fistula and ongoing problems. There was no significant difference between the study group and the control group in terms of QQVCFCA score. However, high type ARM patients were found to have lower QQVCFCA (p <0.001) scores than the control group.

Conclusion: QOL results of high-type ARM patients were significantly higher than low-type ARM. While there is no significant difference in quality of life as total score; In behavior, lifestyle, and depression subtypes, the results were worse than the control group.

Keyword: Anorectal Malformation, Quality of Life, QQVCFCA

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INTRODUCTION

Anorectal malformations (ARM) constitute a group of diseases known for centuries, primarily affecting the urogenital system and other systems. These are rare anomalies, with an average incidence of approximately 1 in 4,000-5,000 live births, as reported in various publications (1-3). While present in both genders, they are slightly more prevalent in There is evidence of familial males. predisposition and genetic susceptibility (4-8). The etiology is uncertain, and it is considered to be multifactorial (9). Fecal incontinence following surgical treatment is a distressing condition that impairs the quality of life, affecting both patients and their families. Despite surgical intervention in ARM patients, they often experience lifelong issues related to defecation (10-13)

In patients with ARM, the sphincter mechanism is variably affected. Striated muscles may be underdeveloped or completely normal. Sensation and proprioception are impacted to varying degrees. Constipation is a significant

issue in patients where the rectosigmoid colonic innervation is preserved during surgery. It is the commonly encountered most problem postoperatively, more frequently observed in patients ARM. with low-type Megarectosigmoid colon may develop due to constipation, leading to further impairment of bowel movement and overflow fecal incontinence. Soiling. a less common manifestation, represents a leakage of stool in a smearing pattern and occurs more rarely than constipation (14).

classifications, Despite numerous the anatomical classification proposed by Stephens and Smith in 1984, known as the Wingspread has been classification, the most comprehensible and widely used until recent times. Based on the relationship of the blind end of the rectum with the levator muscles, this classification divides anorectal malformations into three main groups: high (supralevator), low type (infralevator), and intermediate type. The most common type in males is rectobulbar fistula, while in females, rectovestibular fistula ranks first (Table 1) (15).

Table 1. The Wingspread classification in anorectal malformations (ARM)

	High	Intermediate	Low	Unclassifiable
Female	Fistula-free	Rektovestibula fistula	Anovestibular fistula	Rare
	anorectal agenesis	Rektovaginal fistula	Anocutaneous fistula	malformations
	Rektovaginal fistula		Anal stenosis	Persistan cloaka
Male	Fistula-free anorectal agenesis	Rectrourethral fistula Fistül-free anal agenezis	Anocutaneus fistula Anal stenosis	Rare malformations
		_	Anovestibular fistula	

In anorectal malformations, the overall incidence of associated anomalies is known to

range between 25% and 75% (16). Particularly, genitourinary system anomalies are quite common. While approximately 60% of high

and intermediate-type ARM cases exhibit genitourinary malformations and vesicoureteral reflux, the frequency of genitourinary malformations in low-type ARMs is only about 15-20% (17). Cardiovascular anomalies are also frequent in association with ARM. Anomalies such as ventricular septal defects (VSD) and Tetralogy of Fallot are present in 22% of cases (4, 18). Vertebral anomalies, on the other hand, are more commonly related to the sacrococcygeal region, including conditions like sacral agenesis and spina bifida. Tethered cord is the most common spinal anomaly associated with ARM (19). Gastrointestinal system anomalies also frequently accompany ARM, with esophageal atresia observed in 10% of cases (20).

The primary objectives in the examination, imaging, and laboratory studies for anorectal malformations are to understand the type of atresia, demonstrate any existing fistula, and detect additional anomalies. The success of surgical treatment is closely associated with the presence of concurrent anomalies, the chosen surgical technique, and particularly the innervation status of the anorectal region (21,22).

The PSARP (Posterior Sagittal Anorectoplasty) technique, developed by Pena and de Vries in 1982 and currently employed as the standard surgical approach for ARM treatment worldwide, is recognized for its enhanced

preservation of sphincter muscles and perirectal nerves (23).

Despite the successful implementation of treatment in children with anorectal malformations, issues such as constipation, fecal incontinence, and soiling may manifest in the subsequent period. These problems can lead to consequences that impact the child's social life, school, friendships, home, and family relationships (24-26).

In this study, our primary objective is to investigate the quality of life in patients with ARM who have undergone surgery and completed definitive operations at our clinic, based on ARM subtypes. Additionally, we aim to compare these patients with individuals from a healthy population who experience fecal incontinence due to surgical and non-ARM-related causes. The focus is on assessing and contrasting the quality of life among these groups.

METHODS

The study group consisted of 116 patients aged 4 and above with ARM who underwent surgery and completed definitive operations at the Pediatric Surgery Clinic of University of Health Sciece Izmir Dr. Behçet Uz Children's Diseases and Surgery Health Practice and Research Center between 2004 and 2018. These patients had a minimum of 6 months elapsed since the completion of their definitive operations and did not exhibit motor or mental retardation. The

control group consisted of 12 children who had a fecal incontinence with no organic pathology. The data of control group obtained based on by interviewing the families of 348 patients who had surgery for phimosis in our clinic between 2018 and 2019.

One participant from the study group succumbed, 28 individuals were unreachable, 7 declined to participate despite being reached, and 5 did not attend follow-up appointments at the outpatient clinic; thus, they were excluded from the study. The study was completed with a total of 75 participants with ARM. For the control group, comprising 348 children aged 5-15 who underwent surgery for phimosis without any additional medical conditions, 12 (3.4%) were selected based on complaints of fecal incontinence or soiling.

The assessment of physical examination and administration of the questionnaire involved contacting patients and their families via telephone and inviting them to the hospital. Family members were informed about the study, and informed consent was obtained by signing the informed consent form. Patients were subjected to the Quality of Life in Children and Adolescents with Fecal Incontinence Questionnaire (QQVCFCA), which consists of 24 questions grouped into four categories: lifestyle, behavior, depression, and embarrassment. Each question was scored from negative to positive on a scale of 1-2-3-4. The total scores for each group were calculated by dividing the sum of the scores by the number of questions, resulting in average scores.

In our study, patients with a stoma were classified as having a high-type an ARM, while those without a stoma were considered to have a low-type ARM. The physical examination of patients was conducted with the permission of their families, provided appropriate outpatient clinic conditions were ensured. Patient records were retrospectively reviewed to gather information on the gender, birth dates, age at presentation, operation dates and methods, and any additional anomalies if present.

Ethical approval was obtained with protocol number 2019/272 from the Institutional Review Board.

Statistical analysis

Statistical analyses were conducted using the SPSS (Statistical Package for Social Sciences, IBM Inc., Chicago, Illinois, USA) 22 Windows program for the evaluation of the acquired data. The chi-square test was employed for the analysis of categorical data, the Mann-Whitney U test was used for variables not following a normal distribution, and Kruskal-Wallis oneway analysis of variance was applied for comparing mean and standard deviation values across more than two groups.

RESULTS

The age of participants in the study group ranged from 5 to 20 years, with a mean age of 9.06±2.82 years. Of the study group, 43

(57.4%) were male, and 32 (42.6%) were female. Thirty-five patients (46.7%) were aged 10 or above. Among them, 49 (65.3%) had a low-type ARM, and 26 (34.7%) had a high-type ARM. A total of 28 patients (37.3%) had fistulas, while among those with a high-type ARM, 24 (92.3%) had fistulas.

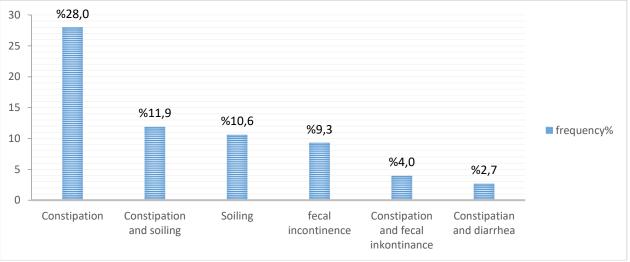
While 53.1% of participants with a low-type ARM were male and 46.9% were female, 65.4% of those with a high-type ARM were

male, and 34.6% were female. The distribution of study group participants based on ARM subtypes and gender is presented.

In the study, patients who was performed colostomy were considered as high-type ARM, while those without colostomy were considered as low-type ARM. Due to different surgeons different clinical approaches varied. Therefore, Table 2 includes patients with rectovestibular fistula in both high and low-type ARMs.

Table 2. Distribution of participants in the study group according to ARM subtypes

	Male	Female	
	Number (%)	Number (%)	Total (%)
Low type ARM			
Fistula-free imperforate anus	14 (87,5)	2 (12,5)	16(32,7)
Anteriorperianal fistula	5 (31,2)	11 (68,8)	16 (32,7)
Rectovestibular fistula	-	8 (100,0)	8 (16,3)
Anocutanezosis fistula	4 (100,0)	-	4 (8,1)
Anal stenozis	3 (100,0)	-	3 (6,1)
Anteriorektopic anus	-	2 (100,0)	2 (4,1)
Total	26 (53,1)	23 (46,9)	49 (100,0)
High Type ARM			
Rectovesikal fisula	6 (100,0)	-	6 (23,1)
Rectobulbar fistula	6 (100,0)	-	6 (23,1)
Rectoprostatik fistula	4 (100,0)	-	4 (15,3)
Rectovestibular fistül		5 (100,0)	5 (19,2)
Cloaka		3 (100,0)	3 (11,6)
Fistula-free rectal atresia	1 (50,0)	1 (50,0)	2 (7,7)
Total	17 (65,4)	9 (34,6)	26 (100,0)



Graph 1. Distribution of postoperative bowel problems among participants in the study group

Table 3. Comparison of QQVCFCA scores between participants in the study group and the control group

		Median	Mean±SD	25 – 75 Percentiles	Test value*;	
OOVCECA	ARM	14,28	13,19±2,75	10,47-15,42	379,500;	
QQVCFCA	Control	14,45	14,56±0,81	13,71-15,49	0,385	
I 'C	ARM	3,42	3,26±0,72	2,71-4,00	241,000;	
Lifestyle	Control	4,00	3,84±0,19	3,61-4,00	0,009	
Dalamia	ARM	3,57	3,42±0,63	3,00-4,00	282,500;	
Behavior	Control	3,85	3,84±0,12	3,71-4,00	0,036	
D	ARM	3,42	3,24±0,64	2,71-3,71	270,000;	
Depression	Control	3,86	3,67±0,22	3,43-3,86	0,026	
Endonesia	ARM	3,67	3,26±0,89	2,33-4,00	337,000;	
Embarrassment	Control	3,33	3,19±0,48	3,00-3,67	0,153	

^{*}Mann Whitney U test

At least one additional anomaly was present in 38 patients (50.7%) with ARM. Upon evaluating ARM patients for associated anomalies in the study, 24 patients (32.0%) exhibited genitourinary anomalies, 20 (26.7%) had cardiac anomalies, 9 (12.0%) presented gastrointestinal anomalies, 8 (10.7%) had skeletal anomalies, and 7 (9.3%) showed neurological problems. Stomas were created in 26 cases (34.7%).

Among the participants with low-type ARM in the study, 21 (42.9%) underwent PSARP, 18 (36.7%) underwent anoplasty, and 10 (20.4%) underwent mini-PSARP. All patients with high-type ARM had a stoma created.

Thirty six patients (48%) reported active complaints related to bowel issues or fecal incontinence. The number of patients with favorable physical examination findings was 43 (57.3%). After surgery, 50 patients (57.3%)

reported experiencing surgery-related bowel problems. The distribution of bowel problems among the participants in the study group is presented in Graph 1.

While 15 patients (30.6%) with low-type ARM reported ongoing bowel problems, 20 patients (80.8%) with high-type ARM reported experiencing continued bowel problems. Among low-type ARM patients, 9 (18.4%) had only constipation, 2 (4.1%) had only fecal 2 incontinence, (4.1%)experienced constipation and soiling, 1 (2%) had fecal incontinence and constipation, and 1 (2%) had only soiling problems. Among high-type ARM patients, 6 (26.9%) had only soiling, 6 (23.1%) had constipation and soiling, 5 (19.1%) had only fecal incontinence, 2 (7.7%) had diarrhea and constipation, 1 (3.8%) had constipation and fecal incontinence, and 1 (3.8%) had only constipation problems.

In the study group, the QQVCFCA score of patients with low-type ARM was found to be significantly higher compared to those with high-type ARM (p<0.001). Additionally, it was observed that the QQVCFCA score was lower in participants with additional anomalies (p<0.001),those with fistula presence (p<0.001), and those experiencing ongoing problems postoperative (p<0.001). Furthermore, in advanced analysis, patients with favorable physical examination findings were found to have a significantly higher QQVCFCA score compared to other patients (p<0.001).

The QQVCFCA (p<0.001), lifestyle (p<0.001), behavior (p<0.001), depression (p<0.001), and embarrassment (p<0.001) scores of patients with low-type ARM were observed to be higher than those of patients with high-type ARM.

No significant difference was found in the median scores of QQVCFCA between those with and without additional anomalies in both high-type and low-type anorectal malformations (p=0.615, p=0.218)

No significant difference was observed in QQVCFCA scores (p=0.385) between the study group and the control group. However, it was noted that the lifestyle (p=0.009), behavior (p=0.036), and depression (p=0.026) scores of ARM patients were lower compared to the control group. Nevertheless, there was no

significant difference in embarrassment scores (p=0.153) between the two groups. The comparison of QQVCFCA scores between the study group and the control group is presented in Table 3.

No significant difference was observed in QQVCFCA scores (p=0.296) between patients with low-type ARM and the control group. However, it was noted that the QQVCFCA scores of patients with high-type ARM were significantly lower compared to the control group (p<0.001).

The median QQVCFCA scores of patients with low-type ARM were higher for those with favorable physical examination findings compared to those with unfavorable findings (p=0.005). In patients with high-type ARM, the median QQVCFCA scores of those with favorable physical examination findings were higher compared to those with both moderate and unfavorable physical examination findings (p=0.003).

DISCUSSION

The incidence of additional anomalies in all anorectal malformations varies between 25-75% (4, 5, 27). According to different series, the frequency of urogenital anomalies has been reported between 28-89% (5, 17, 28-30). Metts et al. found this rate to be between 38.5-50% (31). In our study, genitourinary system anomalies were the most common, occurring in 32% of cases. Generally, cardiovascular anomalies are observed in ARM patients at a

rate of 12-22%. In some series, this rate has been reported between 6-27% (30-33). The frequency of concomitant cardiovascular anomalies in our study was found to be 26.7%.

postoperative complaints When evaluated, it was determined that 36 out of 75 patients (48%) experienced ongoing bowel problems. The most common accompanying problems were constipation (27.8%),constipation and soiling together (22.2%), fecal incontinence (19.4%), and soiling alone (19.4%). In a study by Oyania et al., it was reported that out of 100 patients, 35% experienced ongoing bowel problems, with soiling, 23% 49% having experiencing 39% constipation, and having fecal incontinence (34). On the other hand, in another study by Çavuşoğlu et al., constipation was found in 79% of cases, and fecal incontinence or soiling was observed in 74% of the same patient group (35).

Ongoing bowel problems were present in 16 patients with low-type ARM (32.6%) and 20 patients with high-type ARM (76.9%). The likelihood of experiencing bowel problems increases with the complexity of the deformity. Zheng et al. also hypothesized this and demonstrated fewer complications in patients with low-type ARM in their study (36).

Similar to other related studies, the control group was selected from a healthy population (24, 34, 37). The main reason for our preference for both populations in the study to have fecal

incontinence, as in other similar studies, is to emphasize the impact of ARM on bowel function and quality of life.

In our study, the QQVCFCA score in children under 10 years was found to be lower compared to children over 10 years; however, the result was not statistically significant (p=0.080). Grano's study, similar to our research, identified a positive correlation between increasing age and better quality of life results (24). This outcome may be associated with the growing acceptance of the disease and the gained experience in coping with the problem as children and their families age. In a study by Hartman et al., bowel functions were shown to improve with increasing age, but the quality of life was negatively affected with age, attributed to psychosocial difficulties in adolescence (38). Wigander's study yielded similar results to Hartman's, although statistical significance was not established (39).

The impact of gender on the quality of life (QoL) in children with anorectal malformations (ARM) was not demonstrated (p=0.312). In the study by Grano et al., better QoL results were observed in girls in the child age group, while in the adolescent group, boys showed better results (24). Michel et al.'s study found no gender differences in the child age group, but in the adolescent group, there was a decline in QoL in girls. Hormonal changes, especially

during adolescence, were considered to be influential in this regard (40).

In our study, it was observed that the QoL scores in patients with low-type ARM were higher in four categories, namely lifestyle, behavior, depression, and embarrassment, as well as the total score (p<0.001). In Tannuri's study using the same questionnaire, scores in the lifestyle and behavior sections were slightly higher in high-type ARM, while scores in other sections were higher in low-type ARM; however, all these results were statistically insignificant (37). In a study conducted by Hartman, it was demonstrated that QoL deteriorates in complex ARM cases (38).

When evaluating anal sphincter tone, it was found that patients with a favorable physical examination, fewer postoperative bowel problems, and those who did not undergo reoperation had better QoL outcomes (p<0.001). When assessed separately as high-type and low-type, both groups showed better QoL in individuals with a favorable physical examination (p=0.003, p=0.005).

The study group did not show a significant difference in QQVCFCA scores compared to the control group (p=0.385). However, lifestyle (p=0.009), behavior (p=0.036), and depression (p=0.026) scores of ARM patients were lower than those of the control group. Nevertheless, there was no significant difference in embarrassment scores between the two groups (p=0.153). When evaluating based on ARM

types, there was no significant difference in QQVCFCA scores between patients with lowtype ARM and the control group (p=0.296). However, high-type ARM patients had lower QQVCFCA scores compared to the control group (p<0.001). Grano et al. demonstrated in their study that malformations adversely affected QoL (26). Our study used a different questionnaire (PedsQL), and when compared with a healthy population, it was found that emotional status, social situation, school performance, and total quality of life were worse in children with ARM. Tannuri's study, which used QQVCFCA, revealed negative effects of ARM on total quality of life and all subgroups (p<0.001) (37).

The abundance of patients residing in different cities and the extended time since the operation date, leading to potential changes in contact information, may have limitations in reaching, engaging, and ensuring continuity with the patients. While the requirement for fecal incontinence in the control group distinguishes our study from others, it resulted in a slightly lower number of participants in the control group compared to the patient population. The QQVCFCA scale was chosen as the measurement tool due to its specific development for the pediatric age group and gastrointestinal disorders. Furthermore, the scales' advantages include their easy comprehensibility, absence of medical terminology, and lack of gender-based variations.

CONCLUSION

In conclusion, the QoL in children with lowtype ARM does not vary significantly compared to pediatric individuals in the community without fecal incontinence. However, in children with high-type ARM, both aspects are adversely affected. Nevertheless, we believe that further research in this domain is warranted.

Ethics Committee Approval: The ethical approval for this study was obtained from the Institutional Review Board of Dr. Behçet Uz Pediatric Diseases and Surgery Training and Research Hospital (ethics committee date and number: 2019/272)

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Author Contributions: Concept: VA, ZGT, Design: VA, ZGT, Data Collection and Processing: VA, ZGT, Analysis and Interpretation: VA, ZGT, Writing: VA, ZGT Conflict of Interest: The author declared no conflict of interest.

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