

## Original Article



# Quality of Life and Anorectal Malformations: A Single-Center Experience

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## OPEN ACCESS

Received: Feb 11, 2022

Revised: Apr 6, 2022

Accepted: May 2, 2022

Published online: Jul 6, 2022

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### Conflict of Interest

The authors have no financial conflicts of interest.

## ABSTRACT

**Purpose:** The treatment and long term clinical outcomes of anorectal malformations (ARM) in children have always been the focus of pediatric surgeons. This study aimed at reporting our experience as far as long-term follow-up of ARM in children is concern.

**Methods:** We enrolled patients treated between 1999 and 2019, and established selection criteria to choose appropriate subjects. A validated questionnaire was used to determine long-term quality of life outcomes.

**Results:** Out of a total of 48 patients treated within the study period, 28 were enrolled in this study. Among the latter, more than 35% had at least one long-time complication, and more than 90% had a good lifestyle. Urinary and fecal continence was achieved in more than 95% of the patients using medical devices.

**Conclusion:** This study aimed to bring up new concepts; taking into consideration all aspects of life in patients with ARM, from school life to sexuality, while evaluating fecal and urinary continence. This is essential for the improvement of the skills of the different specialists involved in the management of these patients, and for the implementation of strategies that can improve postoperative function. Most especially, it will also help improve communication between doctors to ensure an adequate transition of these children into adult life.

**Keywords:** Anorectal malformations; Fecal and urinary incontinence; Life style; Paediatric age

## INTRODUCTION

The treatment and long-term clinical results of children with anorectal malformations (ARM) have always been one of the major interests of pediatric surgeons.

Over the past 50 years, advances in embryologic studies and instrumental diagnosis have allowed an increased understanding of the disease, resulting in “better care” for these patients and increased survival [1-3].

Before the 1980s, the outcomes of surgically treated patients were linked to multiple complications. However, with the advent of the posterior sagittal anorectoplasty (PSARP)

procedure, there has been an increase in the success of surgery, especially with a marked improvement in post-operative function [4].

The World Health Organization (WHO) reported that the quality of life is not only related to the absence of disease but also includes physical, psychological, and social status [5].

Although surgical techniques have evolved in recent years, many factors influence health-related quality of life (HrQoL) for ARM patients.

This study aimed to evaluate the HrQoL of patients with ARM treated at our center, compared with the data obtained in the current literature.

## MATERIALS AND METHODS

Patients treated between January 1999 and January 2019 were included in the study. We created the inclusion and exclusion criteria. Patients treated at our center with any type of ARM without associated neurological problems (to avoid bias) were included. Patients who missed the follow-up, did not complete the questionnaire, and did not want to participate in the study were excluded. The study was approved by the Institutional Review Board of Azienda Ospedaliera, Pediatric Fertility lab (2021/PFL 06). Written informed consent was obtained from legally authorized representatives for anonymized patient information to be published in this article.

We considered the type of malformation, associated anomalies, epidemiological data, and familiarity; each patient was contacted on the phone, and a validated quality of life questionnaire survey was conducted.

The validated questionnaire (PedsQL4.0) was offered directly to children aged > or equal to 6 years and parents of children <6 years of age.

Statistical analyses were performed using IBM SPSS software, version 25 (IBM Co., Armonk, NY, USA). For descriptive analysis of all individual categorical variables, absolute frequencies and percentages were calculated. Contingency tables were computed by analyzing the different distributions using Pearson's  $X^2$  (chi-squared) test or Fisher's exact test (for small sample sizes) to explore possible dependence among multiple categorical variables. A  $p$ -value of  $\leq 0.05$  was considered significant for all analyses.

## RESULTS

During the study period, 48 patients underwent surgery (current age range 1–22 years), of whom 30 were men (62.5%) and 18 were women (37.5%). Patients were selected and classified into different diagnostic classes according to the most recent classifications [3].

Among this cohort, seven patients aged three years or less were excluded (language barrier with parents and refusal to use a cultural mediator); ten were not contacted because of invalid telephone numbers; two died before the date of the interview; and one did not participate for

personal reasons. Among the 28 (58%) subjects, there were 18 men (64.3%) and 10 women (35.7%), demonstrating similar participation rates (60% and 55%, respectively) (Table 1).

There were nine (32%) ARMs with recto-urethral fistula (including three recto-bulbar and six recto-prostatic fistulas), six (21%) ARMs with recto-perineal fistula, five (18%) ARMs with recto-vesical fistula, and five (14%) with recto-vestibular fistula. The remaining four were

ARM with recto-vaginal fistula, one case of cloaca, one case of rectal atresia, and one case of anal stenosis.

Regarding the associated malformations, we identified seven cases of ARM with recto-urethral fistula (77.7%), four cases of ARM with recto-perineal fistula (66.6%), three cases of ARM with recto-vesical fistula (60%), three cases of ARM with recto-vestibular fistula (75%), and one case of ARM with anal stenosis (100%). The clinical data are summarized in Table 2.

Based on questionnaire data, we obtained many important results.

**Table 1.** Clinical and malformations distribution

Type of malformation	<10 years	10–20 years	>20 years
Rectal atresia	-	1 M	-
Anal stenosis	9 F	-	-
Recto-perineal fistula	3 F	3 M 1 F	-
Recto-vestibular fistula	1	3	-
Recto-urethral fistula	6	4	-
Recto-vaginal fistula	-	1	-
Recto-vesical fistula	4	1	-
Persistent cloaca	-	1	-

:- not available.

**Table 2.** Clinical data

Type of malformation	School assistance	Extracurricular activities	Social	Home assistance	Fecal continence	Unirary continence
Rectal atresia	No 100%	Yes 100% >3 h/week	Yes 100% >3 h/week	No 100%	100% continent (without device)	100% continent
Anal stenosis*	No 100%	Yes 100% >3 h/week	Yes 100% >3 h/week	No 100%	100% continent (with device)	100% continent (CIC)
Recto-perineal fistula	No 66.7%	Yes 50% >3 h/week	Yes 66.7% >3 h/week	No 66.7%	50% continent	100% continent
	Yes 33.3%	Yes 50% <3 h/week	Yes 33.3% <3 h/week	Yes 33.3%	33.3% incontinent 16.7% soiling (80% device)	10% nocturnal incontinence
Recto-vestibular fistula*	No 75%	Yes 50% >3 h/week	Yes 75% >3 h/week	No 75%	75% continent	100% continent
	Yes 25%	No 50%	Yes 25% <3 h/week	Yes 25%	25% incontinent <2 times/weeks with device	
Recto-urethral fistula*	No 66.7%	Yes 66.7% (66% >3 h/week and 33% <3 h/week) No 33.3%	Yes 55.6% >3 h/week	No 66.7%	11% continent	Yes 80%
	Yes 33.3%		Yes 44.4% <3 h/week	Yes 33.3%	77.8% soiling 11.1% incontinent (>2 times/weeks) (80% with device)	No 20% (10% CIC 30% with medical device)
Recto-vaginal fistula	No 100%	Yes 100% <3 h/week	Yes 100% >3 h/week	No 100%	100% soiling (>2 times/weeks with device)	100% nocturnal incontinence
Recto-vesical fistula*	No 80%	Yes 60% (66.7% >3 h/week and 33.3% <3 h/week) No 40%	Yes 60% >3 h/week	No 60%	20% continent	100% continent (10% CIC)
	Yes 20%		Yes 40% <3 h/week	Yes 40%	40% incontinent >2 times/week 40% soiling (100% 100% use devices)	
Persistent cloaca	No 100%	Yes 100% >3 h/week	Yes 100% <3 h/week	No 100%	100% continent with device	100% continent

CIC: clean intermittent catheterization.

\*Anorectal malformations with other malformations.

### School

All patients reporting their enrollment in a school had established friendly relationships with their classmates and were satisfied with their results (100%; n=28).

Seventy-five percent (n=21) reported no need for teaching or healthcare assistance during school hours, while 25% (n=7) used one or both of these services ( $p<0.05$ ).

These included 1/3 of the patients with recto-urethral fistula (n=3) and recto-perineal fistula (n=2), 25% with recto-vestibular fistula (n=1), and 20% with recto-vesical fistula (n=1). Fisher's exact test reported  $p>0.05$  for each class of ARM. There were no subjects at the university, and none of them were working.

### Leisure, motor activities, and socialization

Sixty-four point three percent (n=18) of the subjects reported doing activities outside of school hours. There were no significant differences between the two genders: males responded positively in 66.6% of cases (n=12) and females in 60% of cases (n=6).

ARM with recto-vestibular fistula (n=2) and recto-perineal fistula (n=3) reported the highest negative response rate, with 50% of patients not engaging in extracurricular activities. Patients with ARM and recto-vesical fistulas responded negatively in 40% of cases (n=2) and in 33.3% of patients with recto-urethral fistulas (n=3).

All other cases were reported to be engaged in extracurricular activities (n=4).

### Extracurricular activities

Eighty-nine percent of patients engaged in extracurricular activities (n=18) were shown to be engaged in sports and/or different activities; two patients were engaged in activities that did not involve physical activity. Of these, one case of cloaca and one case of recto-prostatic fistula were noted.

Among patients who engaged in extracurricular activities, 72.2% (n=13) reported spending a total of at least three hours per week, and the remaining 27.8% (n=5) reported spending at least one to three hours ( $p>0.05$ ).

All subjects reported having socialization for at least one hour per week (100%; n=28). Sixty-four point three percent (n=18) said that they spend at least three hours per week on entertainment (cinema, walking, or shopping).

### Home and daily life

All patients reported living at home with their parents (100%, n=28). Seventy-one point four percent (n=20) reported being self-sufficient in daily activities. The remaining 28.6% (n=8) reported being assisted by their parents in most daily activities (e.g., cleaning or use of continence devices). None of our patients were admitted to a sheltered facility.

### Sexuality

A total of five patients (three men and two women) interviewed having age at least 16 years were investigated according to some items about sexual life and genital disorders.

Four of five patients (80%) reported that they had never shared emotional relationships with their partners. None of the patients interviewed reported ever having had sexual intercourse. The ages at reported menarche were 14 and 15 years, respectively. No menstrual or genital sensitivity disorders have been reported. No erectile dysfunction was reported in men.

### Fecal continence

Thirty-five point seven percent (n=10) of patients reported total continence, 42.9% (n=12) reported episodes of soiling, and absolute incontinence was reported in the remaining six cases (21%) ( $p<0.05$ ) (Table 2).

In summary, we identified a difference only in cases of recto-urethral fistula that reported more episodes of soiling. Among the 28 patients, 20 reported using medical and non-medical devices (71.4%) (e.g., Peristeen, enemas) or medication (e.g., Macrogol) to empty the bowel.

Among the patients using devices, seven had a recto-urethral fistula (78%), five had a recto-vesical fistula (100%), four had a recto-perineal fistula (66%), one had a recto-vaginal fistula (100%), one had a recto-vestibular fistula (25%), one had anal stenosis (100%), and one had cloaca (100%). Specifically, 10 patients (35.7%) used Peristeen, three (10.7%) used diapers, and three (10.7%) used macrogol. The remaining four patients (14.3%) used enemas as needed or performed colic washes.

In addition, 42.9% (n=12) of the subjects reported that they are not suffering from involuntary stool leaks, 35.7% (n=10) reported episodes less frequently than twice per week, and the remaining 21.4% (n=6) subjects reported episodes more than twice per week.

Among the patients with episodes of less than three leaks, it is necessary to add that only one patient had a permanent colostomy.

### Urinary continence

Urinary continence was assessed as diurnal and nocturnal continence (Table 2).

Patients with rectal atresia forms and cloaca were found to be completely continent. In cases of recto-vaginal fistula, we found complete daytime continence but nocturnal incontinence with more than two episodes of leakage per week.

In the forms with recto-urethral fistula, two patients (22% of the specific ARM class) reported regular involuntary daytime leaks (one with recto-bulbar fistula and one with recto-prostatic fistula), while the remaining were continent; four patients (two with recto-prostatic fistula and two with recto-bulbar fistula) had involuntary nighttime leaks.

Only one case of recto-prostatic fistula associated with an occult tethered spinal cord was treated surgically to achieve continence (Mitrofanoff-urinary tract diversion). No additional genitourinary disorders (e.g., micturition pain or recurrent cystitis) have been reported.

## DISCUSSION

ARM are among the most common congenital abnormalities of the large bowel that require surgical treatment [6].

Despite surgical treatment in infancy, many children continue to experience disturbances in bowel function until adulthood, with an adverse effect on the quality of life [7].

As reported by the WHO, quality of life is not only related to the absence of disease but also includes physical, psychological, and social status [5].

HrQoL characterizes the influence of an individual's health on his or her abilities and perception of physical, mental, and social well-being compared to healthy subjects/controls (generic HrQoL) or within the same disease (disease-specific HrQoL) [8,9].

Symptoms such as fecal and urinary incontinence have a significant impact on the social life of patients with ARM, increasing their anxiety and worry [10].

In a recent systematic review of ARMs, the prevalence of long-term (>10 years) problems was 17-77% for fecal incontinence and 22-87% for chronic constipation [11]. On the other hand, different authors have reported the same HrQoL as healthy controls [6].

Several reviews have shown that HrQoL decreases as patients with ARM grow older. During childhood, the main problem is continence. With age, the view of oneself associated with the disease in relation to everyday life, which thus includes work, socialization, and sexuality, becomes predominant [12,13].

With the more frequent use of laparoscopy for treating high forms of ARM, different studies have not found substantial differences in HrQoL [14].

It has been reported that patients with high ARM or associated malformations do not have a reduced HrQoL, probably due to a different mechanism to adjust a new reality in function to accommodate the limitations of their diagnosis, which allows them to control their organic disease, resulting in a higher HrQoL [14].

In our case series, patients with high ARM had good long-term outcomes. This is probably due to the use of devices from an early age (such as colic lavage or intermittent catheterization), with a consequent adaptation to the clinical condition.

Patients in the lower ARM group, who presented forms of fecal or urinary incontinence, also had a low degree of socialization and participated in fewer extracurricular activities.

However, the questionnaire allowed us to verify that despite the ARM, all patients attended school, had a good degree of socialization, and engaged in extracurricular activities. This study has a few limitations, such as, its single-center design and the small number of enrolled subjects. However, we believe it is essential to monitor these patients at long-term during the post-operative period, as a multidisciplinary outpatient clinic has been dedicated with expert gastroenterologists, urologists, pediatric urologists, andrologists, gynecologists, and psychologists. During adolescence, patients are monitored every 6-8 months and after complete pubertal development, they are observed annually. These patients should be followed-up at least once a year until adulthood.

The purpose of this study was to identify and investigate new aspects of patients with ARM in all aspects of life, from school to sexuality to fecal and urinary continence, which can be

crucial for improving the skills of all specialists involved in the management of these patients, implementing strategies to enhance post-operative function, and, most importantly, enhancing their communication with doctors for a successful transition to adulthood.

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