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# Study of intestinal function in anorectal malformations: the role of Bowel management in quality of life

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**Summary.** *Purpose:* We aimed at investigating bowel function in children and adolescents with anorectal malformations (ARMs) and assess their quality of life (QoL), in order to define a personal program of bowel management improving both clinical condition and self-confidence. *Methods:* A total of 55 patients treated for ARM by Posterior Sagittal Ano-Recto-Plasty (PSARP) from 2000 to 2014 were included into the study. The parents of 41 patients answered two telephone questionnaires about the parents' self-efficacy and about QoL (GIQLI). A modified Peña score system was used to evaluate intestinal function. Twenty patients underwent anorectal manometry. After clinical assessment, all patients underwent an individual bowel management program. A median follow-up of 91.3 months was performed. *Results:* A significant inverse correlation was found between Peña score and GIQLI (r:-0.93, p<0.0001). Anorectal manometry paralleled the Peña score, showing an association between megarectum and constipation and soiling subdomains, especially in patients with higher ARM. Patients who carefully followed the bowel management program had significant improvements in both continence (0.93±1.1 vs 0.45±0.9, p=0.0005) and Peña score (4.6±3 vs 3.4±2.5, p<0.0001), which positively affected their self-confidence (100±26.6 vs 110±23, p<0.0001). *Conclusions:* The synergy of different scores and the evaluation of anorectal physiology proved useful to define the bowel management program, which seems to significantly impact both bowel function and QoL, with specific regard to soiling. Moreover, the Peña score might be also quantitatively used, as it parallels with both anorectal manometry and GIQLI, and the latter seems to be suitable for children. Further studies are required to confirm our findings. (www.actabiomedica.it)

**Key words:** anorectal malformations, quality of life, intestinal function, Bowel management, continence, Peña score

## Introduction

Anorectal malformations (ARMs) are a complex group of congenital disorders presenting with an intrinsic poor bowel function, which often causes defecation problems even after proper surgery (1-5). Constipation and soiling represent the most frequent affecting problems of children and adolescents with ARM, with a significant impact on their quality of life (QoL) (5-8). The aim of this study was to investigate the intestinal function in patients with ARM, through clinical scores and anorectal manometry, and to measure their QoL, in order to define an efficient individual bowel management (BM) program. It is still difficult to standardize the clinical assessment of fecal continence, but the widely accepted qualitative

measure is the Peña score system (Krickenbeck protocol 2005) (5, 9, 10). The evaluation of QoL in children and adolescents with ARM is even harder; however, several models of questionnaires are available (11, 12). We intended to prove the essential role of BM in the long-term follow-up of ARMs, improving both clinical conditions and self-confidence.

## Methods

This study included 55 patients treated for ARM by Posterior Sagittal Ano-Recto-Plasty (PSARP) from 2000 to 2014 at our institution (60% male, median age: 8 years). This group of patients is composed of 22 perineal fistulas (40%), 9 vestibular fistulas (16.4%), 8 prostatic recto-urethral fistulas (14.5%), 5 recto-vaginal fistulas (9.1%), 4 cloacas (7.2%) - 3 long-gap (>3cm) - , 3 imperforate anus without fistula (5.5%), 2 bulbar recto-urethral fistulas (3.6%), 1 rectal atresia (1.8%), 1 cloacal exstrophy (1.8%) (Table 1). Among the recto-vaginal fistulas, one is the variant H-type fistula. 14 out of 55 patients showed syndromic association of malformations: 11 VACTERL (Vertebral-Anorectal-Cardiac-TracheoEsofageal-Renal-Limbs), 2 HD (Hirschsprung Disease), 1 Currarino Syndrome. In particular, cardiac malformations affected 13 patients (23.6%), urogenital 12 (21.8%), orthopedic 16 (29.1%).

The majority of patients underwent one-step (32.7%) or three-step (29.1%) surgery and 25.5% two-steps (PSARP with or without protective colostomy). 10.9% underwent more than 4 operations, in this last group there are patients coming from other centers, who needed re-intervention. The median follow-up was of 91.3 months.

The parents of 41 patients answered the AIMAR questionnaire (0-75 points) about the parents' self-efficacy in coping with the ARM. It includes 15 questions, with the parents' personal evaluation of their ability to handle dilatations and hospitalization, and their worry about the future bowel management and QoL of their children (1-5 points per question – not at all/ little/ on average/ very/ totally able to do sth.).

The parents of the patients older than 3 years (N=31) answered the GIQLI (Gastro-Intestinal Quality of Life Index) questionnaire (0-144 points), which is composed of 36 questions (0-4) about the impact of the patients' gastrointestinal disorders, on their mood and everyday life (Table 2). We did not consider question number 26, about sexual life, because of the age of our patients.

The patients underwent a clinical examination with the investigation of defecation habits, urinary and fecal continence, diet, physical activity, relationships with classmates and friends, school performance and other concomitant disorders.

We used a modified Peña score system, grading the qualitative criteria (1-11 points), to measure voluntary bowel movements (1-3), the degree of constipation (0-3), and urinary (0-2) and fecal continence (0-3) for patients older than 3 years (N=31) (Table 3).

**Table 1.** Types of ARM in the group of patients and distribution between genders

| Type of ARM | (N = 55) | | Male (N=33) | | Female (N=22) | |
|---|---|---|---|---|---|---|
| | N | % | N | % | N | % |
| Perineal fistula | 22 | 40.0 | 19 | 57.6 | 3 | 13.6 |
| Vestibular fistula | 9 | 16.4 | 0 | 0 | 9 | 40.9 |
| Recto-urethral fistula | | | | | | |
| • Prostatic | 8 | 14.5 | 8 | 24.2 | 0 | 0 |
| • Bulbar | 2 | 3.6 | 2 | 6.0 | 0 | 0 |
| Recto-vaginal fistula | 5 | 9.1 | 0 | 0 | 5 | 22.7 |
| Cloaca | 4 | 7.2 | 0 | 0 | 4 | 18.2 |
| Imperforate anus without fistula | 3 | 5.5 | 3 | 9.1 | 0 | 0 |
| Rectal atresia | 1 | 1.8 | 1 | 3.0 | 0 | 0 |
| Cloacal exstrophy | 1 | 1.8 | 0 | 0 | 1 | 4.5 |

**Table 2.** GIQLI questionnaire (0-144 points)

*Answer: 0=all of the time 1=most of the time 2=some of the time 3=a little of the time 4=never*

| | |
|---|---|
| **1.** | How often during the past two weeks, have you had pain in the abdomen? |
| **2.** | How often during the past two weeks, have you had a feeling of fullness in the upper abdomen? |
| **3.** | How often during the past two weeks, have you had bloating (sensation of too much gas in the abdomen)? |
| **4.** | How often during the past two weeks, have you been troubled by excessive passage of gas through the anus? |
| **5.** | How often during the past two weeks, have you been troubled by strong burping or belching? |
| **6.** | How often during the past two weeks, have you been troubled by gurgling noises by the abdomen? |
| **7.** | How often during the past two weeks, have you been troubled by frequent bowel movements? |
| **8.** | How often during the past two weeks, have you found eating to be a pleasure? |
| **9.** | Because of your illness, to what extent have you restricted the kinds of food you eat? |
| **10.** | During the past two weeks, how well have you been able to cope with everyday stress? |
| **11.** | How often during the past two weeks, have you been sad about being ill? |
| **12.** | How often during the past two weeks, have you been nervous or anxious about your illness? |
| **13.** | How often during the past two weeks, have you been happy with life in general? |
| **14.** | How often during the past two weeks, have you been frustrated about your illness? |
| **15.** | How often during the past two weeks, have you been tired or fatigued? |
| **16.** | How often during the past two weeks, have you felt unwell? |
| **17.** | Over the past week, have you woken up in the night? |
| **18.** | Since becoming ill, have you been troubled by changes in your appearance? |
| **19.** | Because of your illness, how much physical strength have you lost? |
| **20.** | Because of your illness, to what extent have you lost your endurance? |
| **21.** | Because of your illness, to what extent do you feel unfit? |
| **22.** | During the past two weeks, how often have you been able to complete your normal daily activities (school, work, household)? |
| **23.** | During the past two weeks, how often have you been able to take part in your usual patterns of leisure or recreational activities? |
| **24.** | During the past two weeks, how much have you been troubled by the medical treatment of your illness? |
| **25.** | To what extent have your personal relations with people close to you (family, friends) worsened because of your illness? |
| **26.** | To what extent has your sexual life been impaired (harmed) because of your illness? |
| **27.** | How often during the past two weeks, have you been troubled by fluid or food coming from your mouth (regurgitation)? |
| **28.** | How often during the past two weeks, have you felt uncomfortable because the speed of your slow speed of eating? |
| **29.** | How often during the past two weeks, have you had trouble swallowing food? |
| **30.** | How often during the past two weeks, have you been troubled by urgent bowel movements? |
| **31.** | How often during the past two weeks, have you been troubled been troubled by diarrhea? |
| **32.** | How often during the past two weeks, have you been troubled been troubled by constipation? |
| **33.** | How often during the past two weeks, have you been troubled been troubled by nausea? |
| **34.** | How often during the past two weeks, have you been troubled been troubled by blood in the stool? |
| **35.** | How often during the past two weeks, have you been troubled been troubled by heartburn? |
| **36.** | How often during the past two weeks, have you been troubled been troubled by uncontrolled stools? |

Moreover, 20 patients, over 4 years of age, underwent an anorectal manometry for evaluating rectoanal inhibitory reflex (RAIR), sphincter tone and voluntary squeeze pressure. The examination was managed through a pediatric four channel probe (9Fr-500 mm), ending with a balloon (inflatable channel: 7x200 mm). We measured pressure at 5 and 10 cm above the muco-cutaneous line into the rectum, at rest and during squeeze, and RAIR with 5,10,15,20 ml of air insufflated in 3 seconds. The sphincter tone contractility was registered at 2.5 cm.

Finally, we elaborated a BM program for each patient. We gave dietary suggestions on good hydration and adequate fiber intake. Several patients needed the use of macrogol-based laxatives, whereas others required enemas. We proposed the trans-anal irrigation (Peristeen®) to older children and adolescents with severe constipation or soiling.

After a year of BM program, in February 2015 we re-tested the patients with the modified Peña score system and the GIQLI questionnaire.

**Table 3.** Modified Peña score system (1-11 points)

| Subdomains | Grade | Score |
|---|---|---|
| 1. Voluntary bowel movements | • Feeling of urge | 3 |
| | • Capacity to verbalize | 2 |
| | • Hold the bowel movement | 1 |
| 2. Soiling | • No | 0 |
| | • Occasionally (once or twice per week) | 1 |
| | • Every day, no social problem | 2 |
| | • Constant, social problem | 3 |
| 3. Constipation | • No | 0 |
| | • Manageable by changes in diet | 1 |
| | • Requires laxatives | 2 |
| | • Requires enema | 3 |
| 4. Urinary incontinence | • No | 0 |
| | • Mild dribbling/wetness day and night | 1 |
| | • Complete incontinence | 2 |

## Results

The results obtained from the parents' self-efficacy questionnaire (AIMAR) are ordered in accordance with the degree of assumed ability to face the problem expressed in each question (5 levels: not at all/ little/ on average/ very/ totally able to do sth.) (Fig. 1). A surprising finding was that the parents of children with more complex associations of malformations, felt more able to face difficulties. However, the majority of parents affirmed to be able to manage the child's bowel disorders, and recurrent hospitalizations, even far from home. The main difficulties for the parents referred to anal dilations, combining different medical opinions, lack of adequate services, and, obviously, to the concern about their children's future.

Through the modified Peña score system (0-11), we could measure the degree of constipation and continence as showed in the figure below (Fig. 2). The following analysis refers to the first evaluation at the zero point, before starting the BM program. Patients with a score of 1 had a good bowel and urinary function, and did not need any treatment. Patients with a score of 10 were two complex cases of cloaca, who presented urinary and fecal incontinence.

A more interesting analysis was elaborated di-

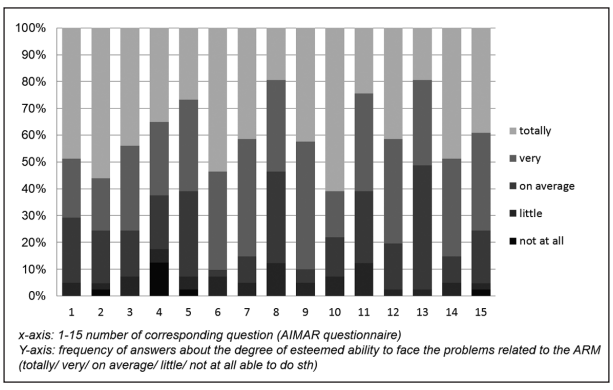


**Figure 1.** Parents' self-efficacy questionnaire results

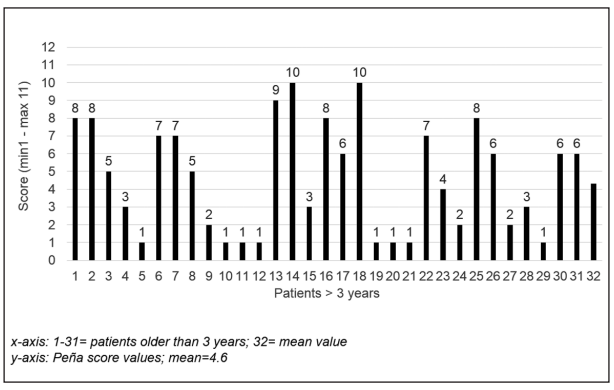


**Figure 2.** Peña score (2014)

viding the patients into two groups: "high" ARMs (H-ARMs) (recto-urethral fistula, cloaca) and "low" ARMs (L-ARMs) (perineal fistula, vestibular fistula, imperforate anus without fistula) (Fig. 3). It was indeed evidenced that fecal incontinence prevailed among H-ARMs, whereas in the other group constipation was the most common disorder (Fig. 3).

In particular, 74% of patients with L-ARMs could hold bowel movement versus 10% of H-ARMs, where the feeling of urge was rather frequent (40%). Soiling was absent (63.2%) or occasional (26.3%) among L-ARMs, whereas it was almost constant in H-ARMs (90%) and often caused social problems (40%). Constipation was the most recurring problem of both L- (63%) and H-ARMs (100%), requiring enemas in almost all patients of the last group (90%). Lastly, urinary incontinence occurred in 3 cases of cloaca (30% of H-ARMs).

At the zero point, the GIQLI results varied from 55 (the worse) to 137 (the better). The average value was of 100. The worse results came from H-ARMs patients. In particular, abdominal pain, constipation, incontinence, poor general conditions and frustration due to the illness, affected most the patients' QoL.

Anorectal manometry, performed in children older than 4 years (N=20), showed a regular voluntary squeeze pressure in all patients. RAIR was normal in 9 patients, altered in 7 and absent in 4. The anal sphincter tone resulted preserved in 6 patients, whereas 14 had a hypotonic sphincter, with a discontinuous contractility in 15 patients. 11 patients presented signs of megarectum.

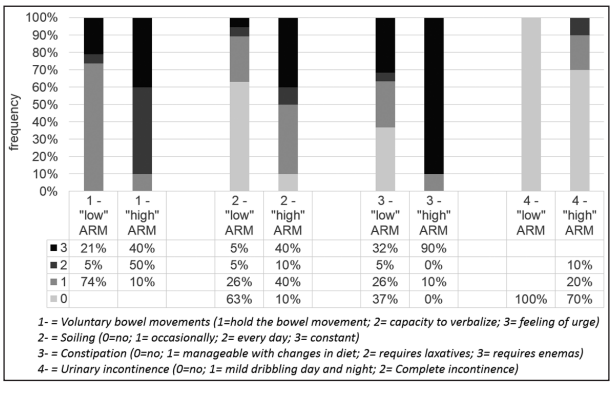


**Figure 3.** Comparison between "low" and "high" ARMs' results of Peña score in the various subdomains (2014)

Distinguishing between H- (N=6) and L-ARMs (N=14), manometry results were different. All H-ARMs had signs of megarectum, altered or absent RAIR and hypotonic sphincter with discontinuous contractility. Among L-ARMs, normal RAIR is prevalent, 9 patients had hypotonic sphincter or discontinuous contraction (64.3% of L-ARM), and just 5 (37.5% of L-ARMs) showed signs of megarectum.

In other terms, anorectal manometry paralleled Peña score, showing an association between megarectum and constipation and soiling subdomains, especially in patients with H-ARMs.

In the end, we calculated the bivariate Pearson correlation between Peña score and GIQLI at the zero point (2014), and we found a significant inverse correlation (r:-0.89, p<0.0001) (Fig. 4).

After a year following the BM program (2015), we re-elaborated the scores and we found once again the same linear significant correlation between Peña score and GIQLI (r:-0.93, p<0.0001) (Fig. 5).

Furthermore, patients who carefully followed the BM program showed significant improvements in both continence (0.93±1.1 vs 0.45±0.9, p=0.0005) (Fig. 6) and Peña score (4.6±3 vs 3.4±2.5, p<0.0001) (Fig. 7), which positively affected their self-confidence and QoL, as showed by GIQLI results (100±26.6 vs 110±23, p<0.0001) (Fig. 8).

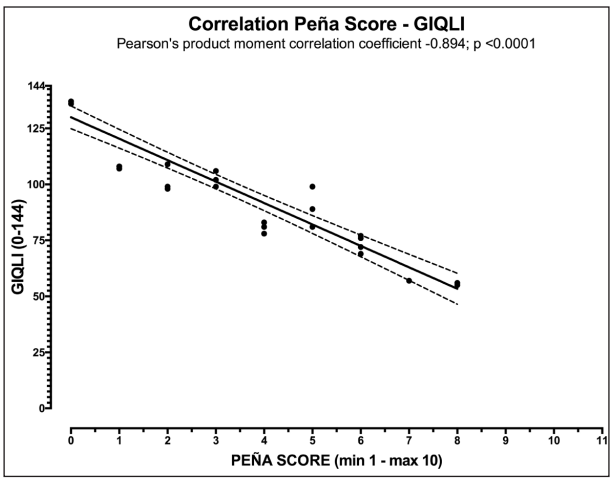


**Figure 4.** Correlation between Peña score and GIQLI at the zero point (2014)

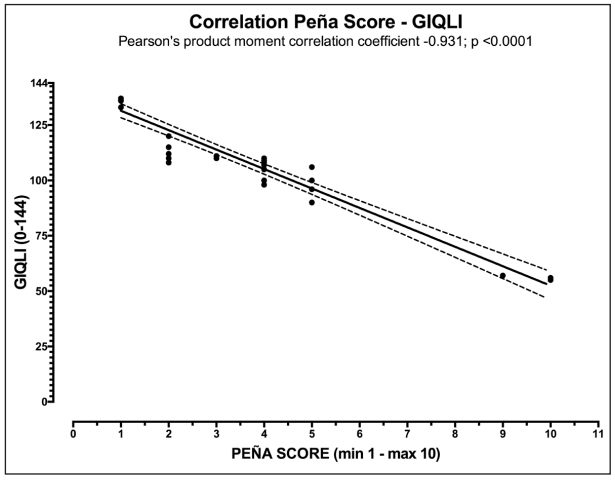


**Figure 5.** Correlation between Peña score and GIQLI after a year of BM (2015)

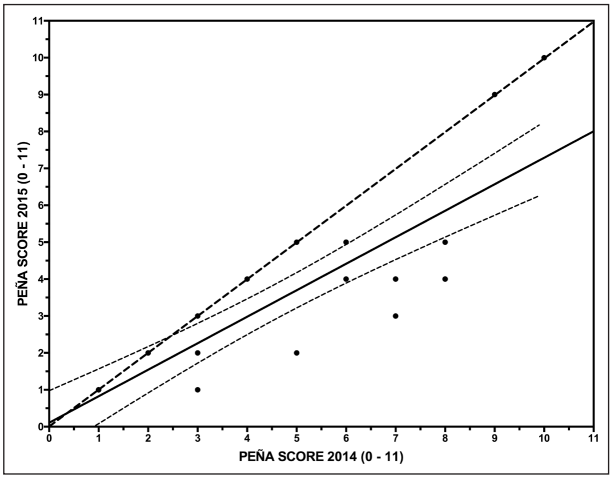


**Figure 7.** Peña score after a year of BM: comparison between 2014 and 2015

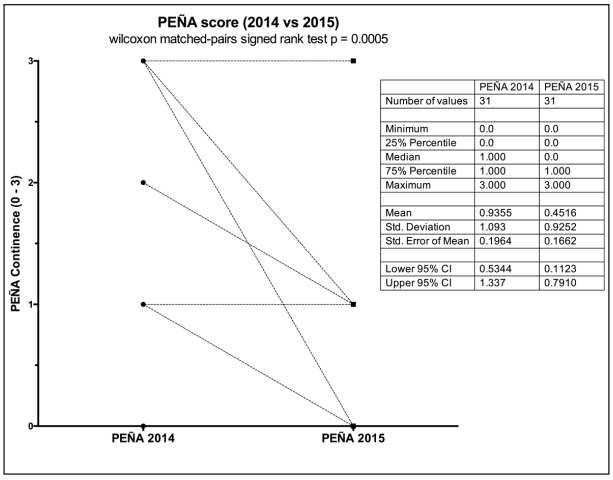


**Figure 6.** Continence subdomain after a year of BM: comparison between 2014 and 2015

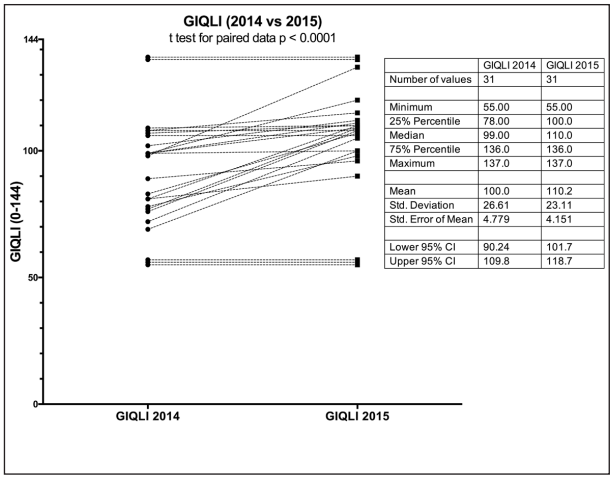


**Figure 8.** GIQLI results after a year of BM: comparison between 2014 and 2015

## Discussions

At the moment, it is yet difficult to measure and compare children's and adolescents' QoL in ARMs, but it is evident that bowel disorders, like chronic constipation and soiling, seriously affect the behavior and the psychological sphere of growing individuals, as occurs with others functional disorders (5, 9, 11, 13). For this reason, we decided to study the intestinal function of patients treated for ARM at our institution, to find the adequate personal BM program to improve QoL. The distribution of types of ARM in our population, in general and between genders, reflects the same trend found in the literature (14). The clinical features and the manometrical data also correspond to the present acknowledgements, in particular with regard to the characteristics that distinguish H- from L-ARMs: a major incidence of fecal incontinence, with a hypotonic sphincter, the first; and severe constipation the last (15, 16). Urinary incontinence affect only two cloaca

patients, who require intermittent catheterization, as usually reported in these cases (1).

Besides the surgical efforts to use the correct technique (minimally invasive approaches could represent a future improvement in this sense) (17-19) also eliminating the last few denervated centimeters of the fistula, there is an intrinsic component of intestinal dysfunction to consider in patients with ARM (20). However, constipation is manageable and, with it, pseudo-incontinence too (21, 22). In most patients, soiling completely disappeared or presented rarely, once following the BM program, which allows the lower intestinal tract to be kept clean. BM is a useful tool also for true fecal incontinence in patients with bad prognosis defects (23-25). This was an important success for our patients, because it changed their social interactions with peers and their ability to do ordinary or recreational activities, beside improving their general physical conditions. BM evolves with the patient, but it often needs to be carried on life-long (26). That is why, the patients' and their parents' compliance is fundamental (27). The parents' seem to be very willing and able to execute the BM program, as the AIMAR questionnaire is evidence of. With regard to the children, when they grow up, they need to understand their condition and to gain independence. For this purpose, the trans-anal irrigation (Peristeen®) proved effective in older children and adolescents (28,29). Therefore, long-term follow-up is mandatory, and the young patients should understand how to use everyday simple expedients to better their condition, even with some psychological help when needed. As we could observe, moreover, compliance enhances after the first positive changes.

This study was useful to demonstrate that, the Peña score can be also quantitatively used and can be a valid instrument for monitoring not simply clinical conditions and response to BM, but also the QoL. Indeed, the score paralleled the results of GIQLI, proving the effective influence of bowel disorders on mood and discomfort. GIQLI expresses the subjective perception of well-being and revealed to be a useful questionnaire for children and adolescents with ARM too, as already demonstrated by other studies (11, 30-32). The patients' evaluation through both the scores at the zero point and after a year of BM is far more interesting: the correlation between Peña score and GIQLI is significant and reflects the clinical improvements in both directions.

## Conclusions

In conclusion, the study of intestinal function in ARMs through clinical instruments and anorectal manometry, and the evaluation of QoL are important to define the proper BM program for each patient, which can significantly improve both general and psychological conditions. Peña score and GIQLI proved useful grading tools for assessing and monitoring fecal and urinary continence, constipation and their consequence on social life. We would carry on our research in a longer follow-up. Further studies are required to confirm our results and to validate QoL measurement systems for patients with ARMs.

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