

Long-term follow-up of anorectal malformation – how long is long term?

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Introduction The ultimate goal of treatment in patients with anorectal malformation (ARM) has moved from mere survival to alleviating symptoms and improving quality of life (QoL), which has become established as an important endpoint in medical care. Adolescents and adults with ARM face several major functional and psychological problems requiring continuity of care.

Materials and methods All patients more than 15 years of age presenting with ARM between June 2010 and 2015 were evaluated. Clinical features, investigations, type of surgery done at birth, present treatment given and outcome were analysed. Anal continence was assessed by Kelly's scoring system. Psychological assessment was done using QoL score.

Results Six male and seven female patients, aged 16–32 years, presented to our institute with problems related to ARM. Two presented with anal stenosis, two with mucosal ectropion, four had constipation and soiling despite adequate opening, two had megarectosigmoid and three had undergone ileostomy elsewhere for distension/obstruction owing to impacted faecoliths during adolescence. Surgical treatment was offered to 10 patients, out of whom two patients on ileostomy refused to undergo any surgery. Patients underwent Malone's antegrade enema procedure (two), ectropion excision (two), revision

anoplasty (two), excision of pouch and anoplasty (two). Psychological assessment showed that patients felt embarrassed and depressed and used lifestyle-coping behaviour. After bowel management and surgery, mean continence and QoL scores improved from 2.7 to 5.1 and 1.26 to 3.04, respectively, which was statistically highly significant ($P=0.001$).

Conclusion Long-term follow-up of patients with ARM is essential to maintain overall QoL. Improvement in continence by bowel management programme and appropriate surgical intervention leads to improvement in QoL. *Ann Pediatr Surg* 14:111–115 © 2018 Annals of Pediatric Surgery.

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Introduction

The ultimate goal of treatment in patients born with anorectal malformation (ARM) has moved from mere survival to alleviating symptoms and improving quality of life (QoL) after surgical reconstruction. Most paediatric surgeons do not get to see the infants that they operated upon for ARM in adulthood. Adolescents and adults with ARM face substantial functional and psychological problems, such as faecal incontinence, soiling, constipation and urological, sexual and psychosocial issues. They face difficulties in coping with these problems or getting correct advice from adult surgeons/physicians.

Over the past 5 years, we encountered patients more than 15 years of age with complications related to previous ARM reconstruction. One measure of the effectiveness of therapy to correct faecal incontinence is the degree to which a patient's QoL is enhanced [1].

Materials and methods

All patients more than 15 years of age presenting with problems related to ARM from 2010 to 2015 were evaluated after approval from the ethical committee. Clinical features, investigations, type of surgery done at birth, present treatment given and results were analysed. Anal continence was assessed by Kelly's scoring system (2 – normal under all circumstances and no soiling, 1 –

occasional escape of faeces or flatus and 0 – no control), staining of underclothes (2 – always clean, 1 – occasional staining and 0 – always stained) and quality of sphincter squeeze (2 – strong and effective, 1 – weak and partial and 0 – none). Psychological assessment was done using QoL score [1] (Table 1), which was assessed before and 1 year after the treatment of presenting complaint. QoL had a set of questions grouped into four scales. Scale response ranged from 1 to 4, with 1 indicating low functional status of QoL. Scale scores are the mean response to all the questions in the scale (add the response to all questions in a scale together and then divide by the number of items in the scale) (minimum score 1, maximum score 4).

Results

A total of 13 patients presented to our institute with problems related to ARM repair at birth and in childhood. Age ranged from 16 to 32 years. One male and one female patient (case no 1 and 12) had no records available about their previous surgery. As per the relatives, both of the patients had undergone three surgeries.

Routine haematological and biochemical investigations were within normal limits. Contrast enema was done for all the patients except one (case 5) who refused for any intervention. Three (cases 1, 2, and 4) had dilated rectosigmoid pouch, and the remaining patients showed

Table 1 Faecal incontinence quality of life scale [1]

Scale 1: Lifestyle

1. I cannot do many things that I want to do
2. I am afraid to go out
3. It is important to plan my schedule
4. I cut down on how much I eat before I go out
5. It is difficult for me to get out and do things like movie
6. I avoid travelling
7. I avoid visiting friends
8. I avoid eating outside
9. I avoid travelling by plane or train
10. I avoid staying overnight away from home

Scale 2: Coping/Behaviour

1. I feel I have no control over my bowel
2. I worry about not being able to get to the toilet in time
3. I try to prevent bowel accidents by being near a bathroom
4. I have sex less often than I would like to
5. Possibility of bowel accidents is always on my mind
6. Whenever I am away from home, I stay near a restroom as much as possible
7. I worry about bowel accidents
8. I can't hold enough my bowel movements to get to the bathroom
9. Whenever I go to new place I specifically locate where the bathroom is

Scale 3: Depression/self-perception

1. In general what would you say your health is (1 – poor, 4 excellent)
2. I feel different from other people
3. I enjoy life less
4. I feel like I am not a healthy person
5. I feel depressed
6. I am afraid to have sex
7. During past month have you felt depressed, discouraged, hopeless or wondered if anything is worthwhile

Scale 4: Embarrassment

1. I leak stool without even knowing it
2. I worry about others smelling stool on me
3. I feel ashamed

Points given for each questions: 1 – most of the time/strongly agree, 2 – some of the time/somewhat agree, 3 – little of the time/somewhat disagree, 4 – none of the time/strongly disagree.

impacted faecoliths with mild dilatation of rectosigmoid. MRI was done for three cases, out of which two showed partial sacral agenesis, absent external sphincter and puborectalis with thinning on left side; the remaining patients were not willing because of the cost factor.

Out of 13 patients, two presented with anal stenosis, two with mucosal ectropion with stenosis, four had adequate opening but constipation and soiling, three had ileostomy and two had megarectosigmoid (owing to impacted faecoliths or congenital). Although they were suffering from these problems for many years, causing psychological discomfort and reduced confidence, they were now mainly concerned about matrimonial and job-related issues (Table 2). Two patients improved on bowel management alone. Ten patients were offered surgical intervention. Two patients on ileostomy refused to undergo any surgery. One of them had undergone total colectomy during her previous surgery by a general surgeon for massive dilatation of colon owing to faecolith impaction (case 5). The other patient (case 1) had undergone three surgeries during childhood and later at 8 years had ileostomy for distension and inability to pass stool. Of the ten patients offered surgery, two underwent Malone's antegrade colonic enema (MACE), two underwent ectropion excision, two underwent revision anoplasty and two underwent excisions of pouch and anoplasty. One female patient had undergone cutback anoplasty at birth for rectovestibular fistula and had normal bowel movements. She delivered a child at

30 years of age, and developed a small tear leading to spillage of faecal matter in her vagina. She had undergone three surgeries for rectovaginal fistula repair through abdominal route at another centre. As no surgery for anal repositioning was done, contrast study again showed spillage into the vagina leading to fallacious impression of rectovaginal fistula. When the anatomy was explained to her, she preferred to have the anal opening in the vestibule as she was not symptomatic (case 7).

Total number of surgeries these patients had undergone from birth until present treatment ranged from 1 to 6. Follow-up of 1 year showed improvement in faecal continence, as well as psychological status (Table 2). The most important desire was to stay clean and the second wish was to lead a normal life without any aided measures (rectal washes, drugs and so on). Marked improvement in the well-being was observed by relatives and care takers the moment faecal soiling disappeared or reduced with aided measures (Fig. 1). Kelly's score and QoL scores improved significantly ($P=0.001$) (Table 3). As the soiling disappeared or reduced, self-perception improved. Out of 10 patients who received treatment, five are continent on bowel management, and five are on laxatives.

Four patients (three male and one female) had urinary incontinence. In the female patient, urinary continence improved after bowel management programme. However, all the male patients required clean intermittent catheterization for neurogenic bladder, two owing to sacral abnormality and the remaining one occurring as a postoperative complication of previous ARM reconstruction. Only one female patient in our study has delivered a child; the remaining six were seeking treatment before marriage. The male patients were concerned about cleanliness, sexual relationship and embarrassment at job. Their fear of embarrassment at job disappeared when they achieved continence with bowel management programme.

Discussion

The type of ARM determines the number of surgeries needed for repair. The long-term functional outcome of patients of ARM depends on anatomy of birth defect and function after the reconstruction [2]. While patients of ARM with mild defect usually develop adequate bowel control, many others have long-lasting problems related to defecation affecting their QoL, which is an important endpoint in medical care [1]. Clinicians should inform, treat and refer their patients to the appropriate caregiver [2].

Paediatric surgeons should be aware of deficits in psychological functioning and therefore not only enquire about their patient's physical well-being but also about their patients' emotional and social well-being. This is especially important in adolescent and adults as they are reported to have lower levels of psychosocial functioning than children [2–4].

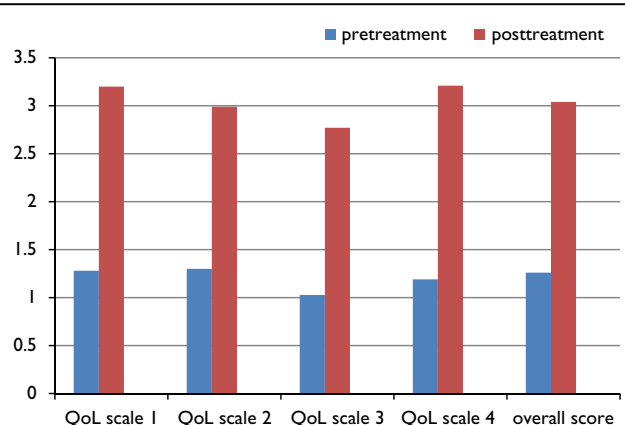
Patients with ARM have to learn to live with a variety of problems, and it remains unclear whether these problems remain consistent over time [2]. In Indian circumstances,

Table 2 Patient profile

Sl. no.	Types of ARM	Previous surgeries	Age at referral (years)/sex	Clinical abnormalities at referral	Complaints at referral	Social reasons for seeking treatment	Surgical intervention after referral	Follow-up (>1 years)	Kelly's score at referral	Kelly's score after intervention	QoL score at referral	QoL score after intervention
1	No details	1. Colostomy 2. No details 3. Colostomy closure 4. Ileostomy at 8 years of age	16/female	Dilated rectosigmoid type IV pouch	Ileostomy	Matrimonial	Refused surgery	Refused	NA	NA	1.34	NA
2	RVF	No surgery for RVF. 1. Ileostomy at 6years of age	18/female	RVF	Ileostomy	Matrimonial	Excision of dilated rectosigmoid and ASARP	Occasional soiling, BM 2–3/day	NA	4	2	3.17
3	RVF	PSARP	22/female	Mucosal ectropion	Constipation soiling	Matrimonial	Ectropion excision	No soiling BM 1–2/day	3	6	1.06	3.17
4	RVF	1. Colostomy 2. PSARP 3. Colostomy closure	23/female	Type IV pouch	Constipation soiling	Treatment	Excision of pouch and pull through	No soiling	2	5	1.37	3.2
5	RVF	No surgery for RVF 1. Colectomy with ileostomy at 18 years of age	23/female	Status ileostomy	Ileostomy	Matrimonial	Refused treatment	–	–	NA	NA	NA
6	RVF	1. Colostomy 2. PSARP 3. Colostomy closure	24/female	Anal stenosis	Constipation soiling Urinary incontinence	Treatment	Anoplasty Bowel management	No soiling	3	6	1.03	3.3
7	RVF	1. Cutback anoplasty at birth 3 Surgeries for rectovaginal fistula after delivery at 30 years of age	32/female	Rectovaginal fistula	Stools in vagina	Family issues	EUA and counselling	–	4	NA	NA	NA
8	Rectourethral fistula	1. Colostomy 2. PSARP 3. Colostomy closure	16/male	Mucosal ectropion	Soiling	School	Ectropion excision	No soiling	3	6	1.1	2.89
9	Rectovesical fistula	1. Colostomy 2. Pull through 3. Colostomy closure	18/male	Sacral hemivertebra	Constipation soiling Urinary Incontinence	Job matrimonial	MACE Bowel management CIC	Occasional soiling BM 2–3/day	2	4	1.34	3.1
10	Rectourethral fistula	1. Colostomy 2. PSARP 3. Colostomy closure	20/male	Anal stenosis	Constipation Soiling	Job	Anoplasty	No soiling, BM 1–2/day	3	6	1.1	2.96
11	Rectourethral fistula	1. Colostomy 2. PSARP 3. Colostomy closure	20/male	Anal opening adequate	Constipation soiling	Job and matrimonial	Bowel management	No soiling, BM 1–2/day	3	5	1.31	3.13
12	No details	1. Colostomy 2. No details 3. Colostomy closure	22/male	Anal opening lax, sacral hemivertebra	Constipation Soiling Urinary incontinence	Job	Bowel management CIC	No soiling, BM 2–3/day	2	4	1.24	3.03
13	Rectovesical fistula	1. Colostomy 2. Pull through 3. Colostomy closure 4. Recurrent Rectourethral fistula repair at 10 5. Recurrent Rectourethral fistula at 12 years of age	23/male	Anal opening adequate	Urinary and faecal incontinence, Using pads	Matrimonial	MACE CIC	No soiling BM 1–2/day	2	5	1.13	2.48

ASARP, anterior sagittal anorectoplasty; BM, bowel movements, CIC, clean intermittent catheterization; MACE, Malone's antegrade colonic enema; NA, not accessed; PSARP, posterior sagittal anorectoplasty; RVF, rectovestibular fistula.

Fig. 1



Pretreatment/post-treatment quality of life scores.

Table 3 Pretreatment/post-treatment quality of life and Kelly's score

Scores	Pretreatment (mean)	Post-treatment (mean)	P value
QoL scale 1: lifestyle	1.28	3.2	0.001
QoL scale 2: coping/behaviour	1.3	2.99	0.001
QoL scale 3: self-perception	1.02	2.77	0.001
QoL scale 4: embarrassment	1.19	3.21	0.001
QoL overall	1.26	3.04	0.001
Kelly's score	2.7	5.1	0.001

QoL, quality of life.

probably parents of patients having a colostomy are initially anxious that their child is not passing stool through the normal route.

Therefore, when reconstructive surgery is done, they heave a sigh of relief. Some amount of incontinence or soiling in the initial phase does not bother them, or they wait in the hope that this will improve with time. However, as the child enters into adolescence the psychosocial problems increases, and in adulthood job and matrimonial issues cause anxiety and low self-esteem. In this scenario, the previous treating surgeon is usually not available.

It is difficult to classify faecal continence. Frequency and consistency of stool, amount of uncontrolled loss, rectal sensation, possibility of holding back defecation, discrimination between formed, loose or gaseous stool and need for therapy are considered to be important factors defining faecal continence [5].

It is important to know and understand whether the adults are content with their status or continent after the repair of high ARM. In a long-term study in patients aged 18 years and older, all patients operated on for high ARM had some form of incontinence. Normal faecal continence was taken as producing faeces once or twice a day of normal consistency at the proper time and place without soiling in between and without taking dietary measures, anal aids or medicine. Nobody could fulfil these criteria, although most patients (84%) were satisfied with the achieved level of

continence with some measures such as drugs, dietary or anal aids [5]. This study concluded that adults after the ARM repair although content (satisfied) were not continent as per the criteria mentioned.

Chronic difficulty in defecation affects the QoL. Most studies found a positive association between disease-specific functioning and QoL, directing attention towards alleviating symptoms in improving psychosocial functioning [4,6,7]. Relationship between disease-specific functioning and QoL remains unclear [2]. Although adults are reported to have lower level of psychosocial functioning [8,9], a few studies reported less faecal problems with adults [9–11]. In our study, patients showed improvement in QoL scores and better self-esteem the moment soiling disappeared. Overall the scores almost doubled (mean: 1.26–3.04 for QoL and 2.7–5.1 for Kelly's score).

Another important goal of management is the preservation of sexual and urological function. There seems to be a close relationship between psychosocial development, urological function and sexual activity. Analysis of 55 patients, 18–56 years of age, showed that 35% of females and 69% of males lived alone. Twenty-six per cent of females became pregnant and 32% males fathered children [12]. Twenty-one patients suffered mucosal prolapse, 18 had megasigmoid/megacolon, 17 had anal stenosis, 14 had permanent neurogenic bladder dysfunction, 23 had recurrent urinary tract infection, 37 patients had to be reoperated and 41 patients needed means of aftercare to achieve social continence [13].

Urinary incontinence may improve with just bowel management as seen in our female patients or require clean intermittent catheterization when secondary to a neurogenic bladder. In our study, only one female had delivered a child. The remaining six were seeking treatment before getting married. Males were concerned about cleanliness, sexual relationship and embarrassment at job. The fear of embarrassment at job disappeared when they achieved continence with bowel management programme. Two patients were happy with MACE and felt confident being independent of relatives for bowel washes. One patient is still requiring rectal washes but is not willing to undergo another surgery for MACE.

Probably these were the few patients who approached various clinicians in the hope of improving their QoL. There might be many others who have exhausted their resources and have lost hopes of improvement or are in depression.

Long-term follow-up of patients of ARM is essential, which might be lifelong at times for some patients. It is important to establish an association or a forum with adult clinicians, so as to have a transition of care and improve their psychosocial, sexual and colorectal functional problems.

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Conflicts of interest

There are no conflicts of interest.

References

- 1 Rockwood TH, Church JM, Fleshman JW, Kane RL, Mavrantonis C, Thorson AG, *et al.* Fecal incontinence Quality of Life Scale: quality of life instrument for patients with fecal incontinence. *Dis Colon Rectum* 2000; **43**:9–16.
- 2 Hartman EE, Oort FJ, Aronson DC, Sprangers MA. Quality of life and disease specific functioning of patients with anorectal malformations or Hirschsprung's disease: a review. *ADC* 2011; **96**:398–406.
- 3 Goyal A, Williams JM, Kenny SE, Lwin R, Baillie CT, Lamont GL, *et al.* Functional outcome and quality of life in anorectal malformations. *J Pediatr Surg* 2006; **41**:318–322.
- 4 Hartman EE, Oort FJ, Visser MR, Sprangers MA, Hanneman MJ, de Langen ZJ, *et al.* Explaining change over time in quality of life of adult patients with anorectal malformations or Hirschsprung's disease. *Dis Colon Rectum* 2006; **49**:96–103.
- 5 Hassink EA, Rieu PN, Severijnen RS, vd Staak FH, Festen C. Are adults content or continent after repair for high anal atresia? A long term follow up study in patients 18 years of age and older. *Ann Surg* 1993; **218**:196–200.
- 6 Rintala R, Mildh L, Lindahl H. Fecal continence and quality of life for adult patients with an operated high or intermediate anorectal malformation. *J Pediatr Surg* 1994; **29**:777–780.
- 7 Diseth TH, Emblem R. Somatic function, mental health, and psychosocial adjustment of adolescents with anorectal anomalies. *J Pediatr Surg* 1996; **31**:638–643.
- 8 Funakosi S, Hayashi J, Kamiyama T, Ueno T, Ishii T, Wada M, *et al.* Psychosocial liaison-consultation for the children who have undergone repair of imperforate anus and Hirschsprung disease. *J Pediatr Surg* 2005; **40**:1156–1162.
- 9 Amae S, Hayashi J, Funakosi S, Kamiyama T, Yoshida S, Ueno T, *et al.* Postoperative psychological status of children with anorectal malformation. *Pediatr Surg Int* 2008; **24**:293–298.
- 10 Stolk EA, Busschbach JJ, Vogels T. Performance of the EuroQol in children with imperforate anus. *Qual Life Res* 2000; **9**:29–38.
- 11 Poley MJ, Stolk EA, Tibboel D, Molenaar JC, Busschbach JJ. Short term and long term health related quality of life after congenital anorectal malformations and congenital diaphragmatic hernia. *Arch Dis Child* 2004; **89**:836–841.
- 12 Schmidt D, Winter S, Jenetzky E, Zwink N, Schmiedeke E, Maerzheuser S, *et al.* Sexual functions in adults with anorectal malformation: psychosocial adaptation. German Network for Congenital Uro-REctal malformations (CURE-net). *Pediatr Surg Int* 2012; **28**:789–792.
- 13 Schmidt D, Jenetzky E, Zwink N, Schmiedeke E, Maerzheuser S. Postoperative complications in adults with anorectal malformation: a need for transition. German Network for Congenital Uro-REctal malformations (CURE-net). *Pediatr Surg Int* 2012; **28**:793–795.