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Original Research Article

Clinical profile, management and post operative outcome in patients with anorectal malformations

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ABSTRACT

Background: Anorectal malformations (ARM) form a very important group of anomalies dealt by pediatric surgeons. Identification of different forms of the anomaly, planning investigation and definitive surgical procedure to attain the final goal of a continent child, remains a difficult task till date. Aim of the study was to study complete clinical profile of the ARM patient, surgical procedure and post operative outcome with special emphasis on continence.
Methods: We conducted a study at Geetanjali medical college, Udaipur, Rajasthan, India for clinical profile and management of patients with ARM with assessment of post-operative outcome. All cases of ARM operated from August 2016 to June 2021 were included in the study. All aspects of the anomaly i.e., type, age and sex distribution, associated anomalies, surgical technique used and post op complications were assessed. Evaluation of continence was done by Kelly's scoring system.
Results: The 71% patients had staged surgery with posterior sagittal ano-rectoplasty (PSARP) being the most common procedure used (71%). Immediate post op complications were nil. Incontinence was present in 29% patients. Constipation was present in 46% patients. On assessing Kelly's scoring 96% patients had satisfactory score. Postoperative continence was achieved in 70.8% and in 83.3% patients after treatment (bowel management).
Conclusions: Meticulous planning and surgical technique with sphincter identification and proper placement of ano-rectum are most important to achieve desired results. Kelly's scoring is important to assess post operative results and plan management for incontinence. With aggressive bowel management it is possible to achieve reasonable continence and quality of life for even those anomalies with poor prognosis.

Keywords: ARM, PSARP, Kelly's score, Fecal incontinence

INTRODUCTION

Ano-rectal malformations (ARM) form a very important group of anomalies dealt by pediatric surgeons. Identification of different forms of the anomaly, planning investigations and definitive surgical procedure to attain the final goal of a continent child, remains a difficult task till date.[1]

Aim of this research was to study complete clinical profile of the ARM patients, management including surgical procedures used and assessment of post - operative outcome with special emphasis on fecal continence.

METHODS

We conducted an observational, descriptive clinical study at our institute (tertiary care center) wherein all cases of ARM operated at Geetanjali medical college (Udaipur, Rajasthan, India), from Aug 2016 to June 2021 were included. All cases were operated and followed up by a

single surgeon. All cases of ARM were included in our study and only those patients were excluded whose case records were incomplete/lost during the course of the study. The study was conducted after taking consent of parents/caretakers in every case and was approved and cleared by the ethical committee of our institute. Data was collected by patients' hospital records, regular post-op OPD visits, questionnaire (proforma with Kelly's scoring chart) and clinical examination of the child. We used SPSS (Statistical package for social sciences) software, 2022 version for statistical analysis. We followed standard management protocol for all patients as given in algorithms (Figure 1 and 2).[2] Our timing of definitive surgery was different as mentioned later. Neonates with ARM were initially stabilized with IV fluids, Injection cephalosporin and metronidazole, investigated as per protocol and followed by anoplasty/minimal PSARP (for low ARM) and colostomy (for high ARM and complex low ARM).

Protocol/algorithm for males

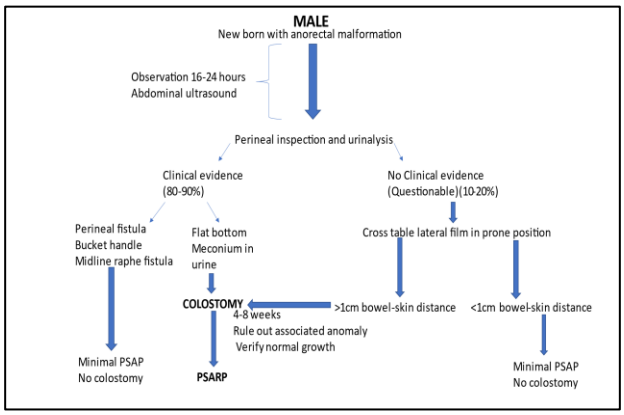

Figure 1: Protocol and algorithm of management-male.[2]

Protocol/algorithm for female patients

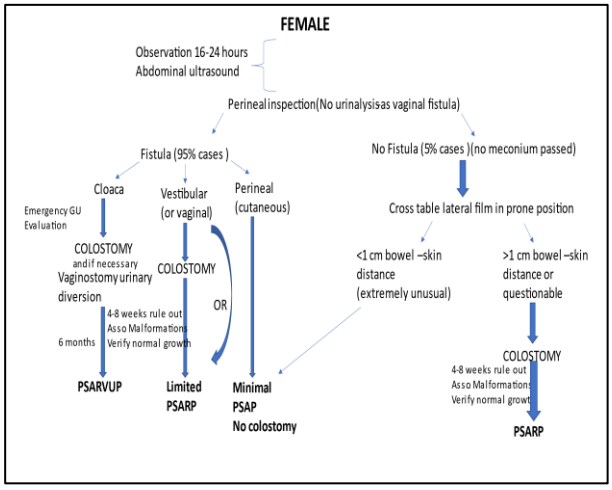

Figure 2: Protocol and algorithm of management-female.[2]

All aspects of the anomaly- type, age and sex distribution, associated anomalies, surgical technique used, post operative complications (immediate and late) were considered. Evaluation of fecal continence was done by Kelly's scoring system. All patients underwent standard investigations i.e., Invertogram/lateral cross table X-ray (for neonates-Figure 3), X-ray abdomen (for colon and spine), USG abdomen (mainly w. r. t. urinary tract anomalies), echocardiography and a mandatory colostogram (Figure 4) before staged PSARP.

Figure 3: Cross table lateral prone x-ray (CTLP), it shows "high" anomaly with rectum (arrow) and marker at future anal site.

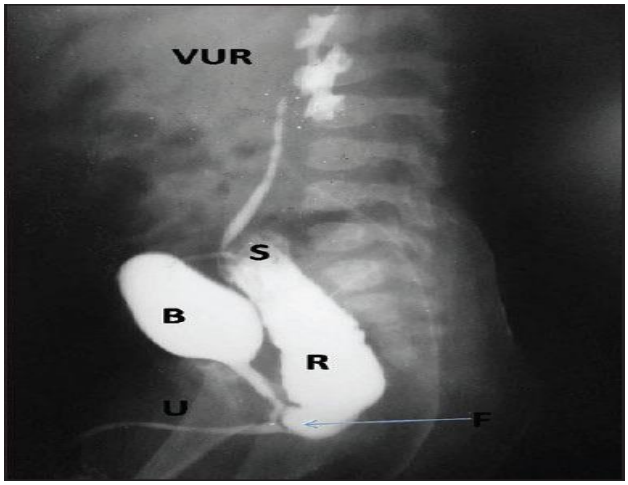

Figure 4: Distal colostogram of recto-bulbar fistula (arrow).

R: rectum, S: sacrum, B: bladder, U: urethra. Also shows an associated vesico-ureteric reflux (VUR).

Pena's PSARP was the standard procedure in majority of cases (Figures 5-7).[3] ASARP (Anterior sagittal ano-rectoplasty) was done in a few cases as required. Primary repair (without colostomy) was done in low ARM and several cases of vestibular fistulas (high ARM in females). For low ARM cases, Anoplasty/minimal PSARP was the procedure done. Staged surgery (with colostomy) was done in cases with (1) Vestibular fistulas where full and satisfactory bowel preparation could not

be achieved, (2) complex low ARMs and (3) All remaining cases (high anomalies). For bowel preparation (in primary procedures), we used polyethylene glycol (pegleg solution) in the dose of 25 ml/kg/hour for four hours and more if required. All patients received inj. cephalosporin, amikacin and metrogyl post-operatively for at least 5 days.

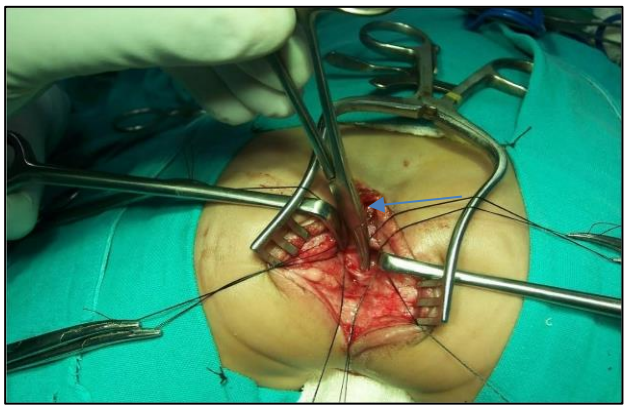

Figure 5: Artery forceps (arrow) demonstrating site of fistula.

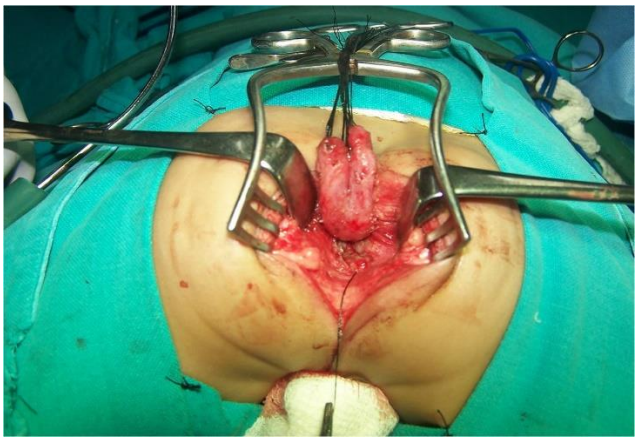

Figure 6: Mobilized rectum.

Figure 7: Neo-anus formed (completed anoplasty).

All patients had anal dilation procedure by appropriate size Hegar's dilator after 3 weeks of PSARP and continued for 6 to 12 months. Colostomy, if present was closed at 2-3 months post PSARP. All patients had a follow-up of minimum of one year after colostomy closure. Patients were called for follow-up at 1 month, 3 months, 6 months and 1 year subsequently and required data collected. Follow-up of many patients was much longer.

Kelly's scoring system was used for continence assessment. It awards points for three basic parameters as given in chart below. An overall score of 5-6 is considered good, 3-4 fair, and 0-2 poor.[4]

Table 1: Kelly's scoring.[4]

Continence	Score
Normal, no soiling	2
Occasional accidents, faeces/flatus escape	1
No control, frequent accidents	0
Staining	
Always clean	2
Occasional staining	1
Always stained	0
Sphincter	
Strong and effective squeeze	2
Weak and partial squeeze	1
No contraction	0

Other complications considered were wound infection, dehiscence, prolapse, stenosis etc.

Patients who had constipation and/or incontinence were treated by aggressive "bowel management programme", discussed later.

Certain terms need to be clarified or defined-1. Incontinence- Involuntary passage of full fecal load or continuous non-stop passage of stool. 2. Soiling- Passage of small amount of fecal matter occasionally in under-garments with otherwise timely/voluntary passage of normal amount of feces. 3. Constipation- It is a bowel dysfunction that makes bowel movements infrequent or hard to pass. In severe cases, it may lead to overflow incontinence.

Above conditions may co-exist. For final consideration/tabulation, we have taken the complications of incontinence and soiling, under a single heading- "fecal leak" for simplicity, as the underlying causes and management for both are similar.

RESULTS

Majority of patients were in the age group of 0-2 years (88%). Median age for PSARP was 12 months.

Table 2: Distribution of male and female population according to age at PSARP, (n=24).

Age (Years)	Male, n (%)	Female, n (%)	Total, n (%)
0-2	11 (46)	10 (42)	21 (88)
2-5	0	1 (4)	1 (4)
>5	0	2 (8)	2 (8)

Table 3: Distribution of type of anomaly male.

Type of anomaly		N (%)
Low	Perineal fistula (low) bucket handle variety	4 (36)
High	Recto bulbar fistula	2 (18)
	Recto prostatic fistula	2 (18)
	Recto vesical	2 (18)
	Pouch colon (Complex variety)	1 (10)

Table 4: Distribution of type of anomaly-female.

Type of anomaly		N (%)
Low	Anterior placed (Ectopic) stenosed anus	1 (8)
High	Vestibular fistula	11 (84)
	"H" type (Rectovaginal) fistula	1 (8)
	Cloaca (Complete)	0

Total cases of low ARM in males were 4 (36%) and high ARM was 7 (64%).

Total cases of low ARM in males were 4 (36%) and high ARM was 7 (64%) (Table 3).

Total cases of low ARM in female was 1 (8%) and total number of high ARM were 12 (92%) (Table 4).

The most common associated anomaly was genitourinary, 11 (45.8%) out of which absent kidney and hydro-ureteronephrosis were most common (each 8.3%). CVS anomalies were present in only 2 patients (8.3%), both male and none required intervention. Skeletal anomalies were not common. No case of spina-bifida or limb deformities was encountered. Sacral deformities (< 3 sacral vertebrae) were found in 4 patients (3 male and 1 female). Overall anomalies more common in males (64%) than females (36%).

Majority 17 (71%) underwent staged surgery. 7 (29%) had primary (without colostomy) surgery. Patients with primary surgery were 2 males (both with "bucket handle" variety of low anomaly) and 5 females (one with anterior placed stenosed anus, and four with vestibular fistula).

Rate of constipation was much higher in recto-bulbar and recto-prostatic fistula whereas it was much lower (zero) in recto-vesical fistula vis -a- vis other study.

Most common were vestibular and recto-vesical fistula, 3 in number followed by one case of recto prostatic fistula and another case of pouch colon.

Table 5: Distribution of study population according to systems involved in ARM.

System involved in ARM		No. of cases involved in our study, n (%)	Males, n (%)	Females, n (%)
Genito-urinary	Total (overall)	11 (45.8)	7 (64)	4 (36)
CVS	Total	2 (8.3)	2 (18)	0
Skeletal (sacrum <3 bodies)	Total	4 (17)	3 (27)	1 (7.7)
Spina bifida/ neural tube defects	Total	0		
Gastro-intestinal	Total	0		

Table 6: Distribution of our study according to type of procedure in ARM.

Type of procedure	N (%)
PSARP (Posterior sagittal anorectoplasty)	17 (71)
ASARP (Anterior sagittal anorectoplasty	3 (13)
Minimal/limited PSARP	2 (8)
Anoplasty	2 (8)

Table 7: Incidence of constipation in different common anomalies and comparison with another large study.

Type of anomaly (list includes male and female anomalies)	Total no. of cases in study	No. of cases with constipation	Out of total cases of same anomaly, (%)	In a study by Neil (%)[5,6]
Perineal fistula(low)	5 (4 male 1 female)	1 (mild) 1 female	20	28.6
Recto-bulbar fistula	2	2 (1 severe 1 mild)	100	55
Recto-prostatic fistula	2	1 (severe)	50	41
Recto-vesical fistula including pouch colon	3	0	0	18.2

Continued.

Type of anomaly (list includes male and female anomalies)	Total no. of cases in study	No. of cases with constipation	Out of total cases of same anomaly, (%)	In a study by Neil (%)[5,6]
Vestibular fistula	11	7 (5 mild 2 severe)	63.6	61.4
Rare (vaginal fistula "H" type fistula	1	0	0	25 (for vaginal fistula)
Total	24	11	45	

Table 8: Incontinence (Fecal Leak) distribution w.r.t type of anomaly and comparison with other large study.

Type of anomaly (list includes male and female anomalies)	Total no. of cases	Cases with incontinence	W. r. t. total cases of specific anomaly (%)	In a study by Neil (%).[5,6]
Perineal fistula(low)	5	-	0	0
Recto-bulbar fistula	2	-	0	31
Recto-prostatic fistula	2	1	50	43
Recto-vesical fistula including pouch colon	3	3	100	82
Vestibular fistula	11	3	27.2	13
"H" Type	1	-	-	Not available
Cloaca	0	-	-	100
Total	24	7	29	57

DISCUSSION

Our study yielded some interesting results. Number of females was more than males. This was different from other studies.[7,8] Percentage distribution of different types of anomalies was similar to other studies done.[9-12] The most common anomaly in males was perineal fistula-bucket handle variety (36%). In female's vestibular fistula (84%) was the most common.

For associated anomalies, the overall (total) number of cases was similar to other studies, but the distribution was different as shown in Table 5. In studies done by Watanabe et al and Gangopadhyay et al most common anomalies reported were genitourinary anomalies (40-50%) followed by cardiovascular (30-35%) and gastrointestinal anomalies (5-10%).[11,13] In our study these were 46%, 8.3% and 0% respectively. Major skeletal anomaly- spina bifida with/ without meningomyelocele was not found in any case in our study. Cases of sacral anomaly (sacrum <3 vertebral bodies) alone were 4 (17%) which was similar to studies by Bhatnagar and Mittal et al.[1,9] CVS anomalies were lower than other studies with only 2 (8.3%) cases of small VSD. No case needed intervention. Whereas, in a study by Bhatnagar, associated CVS anomalies such as VSD, TOF, TGS, were 10-20% etc.[1] This may represent an overall trend in the population with more antenatal diagnosis and termination of pregnancy being done nowadays for major cardiac defects. Gastro intestinal anomalies were not found in our study. Genitourinary anomalies were similar to other studies and were seen more commonly in higher anomalies as expected (Table 5).[1,9]

Coming to post-operative problems, immediate surgical complications like infection, dehiscence, stenosis, prolapse etc. were not found in any of our patients. This was a very satisfactory outcome. An important and frequent long-term problem is constipation. For simplicity we have taken cases of different grades of constipation together. Though theoretically it should be seen more in low lesions due to a large rectal volume and preservation of internal sphincter, it frequently supervenes in both low and high anomalies.[14,15] Among higher anomalies, vestibular anus cases are especially prone to constipation for yet unknown reasons. Interestingly, in our study (Table 7), lower anomalies did have constipation but was lesser as compared to other studies.[5] Rate of constipation was much higher in recto-bulbar and recto-prostatic fistula whereas it was much lower (zero) in recto-vesical fistula vis -a- vis other study[5]. Constipation if severe can lead to over distended rectum loaded with feces and further to "overflow incontinence", as was seen in 3 of our cases with vestibular fistula. The problem can be present in both sexes. Rate of constipation was similar in both studies regarding vestibular fistulas and was predominantly "mild" in our study. None of the male patient with low ARM had constipation (rate=0 %). Overall rate (of total cases) was similar (Table 7).

Constipation has to be aggressively treated as neglecting it can lead to overflow incontinence and worsen to a megarectum or mega-sigmoid which makes it very difficult to treat. These cases even sometime need surgery (resection of severely dilated part or similar to anterior resection in adults. We treat these patients incrementally and as required with diet, laxatives, suppositories, enema or combination of them. Most patients respond well to these measures. Study done by Schmiedeke et al in 10 post operative patients, constipation was found in 6 patients as a long-term problem out of which 2 required dietary modifications, 1 patient required laxatives and 3 patients required enema.[16]

Next and even more important post-op problem is incontinence. The purpose of entire management of ARM cases is not only to give an opening for feces at the normal position of anus but also to ensure the child has normal bowel movements, control over voiding and have no/minimal/very occasional incidence of soiling (i.e., continence). Post operative assessment with Kelly's scoring system form an important part of our study. We found that very large number of patients had satisfactory results in 96 % cases (good and fair score together), good in 17 patients (71%) and fair in 6 patients (25%). Only one patient (4%) was found to have poor score. As will be discussed later even this case had a very satisfactory outcome after necessary intervention. Results and comparative study are summarized in table 8. This was better than a similar study by Tsuji et al.[17] Patients with fecal leak had Kelly's score of less than equal to 4. Total number of patients was 7(29%). Out of which 3(42.8 %) were female patients and 4(57.14 %) were male patients. Most common were vestibular fistula, 3 in number each with a Kelly's score of 4, followed by recto-vesical fistula which were 2 in number one with the score of 3 and other one with score of 1. There was one case of recto prostatic fistula with a score of 3 and another case of pouch colon with score of 4. Similar findings were seen in a study done by Bliss et al.[18]

In our study the distribution of fecal leak in different anomalies according to anal/sphincter tone was similar to a study done by Iwai et al with higher anomalies showing weaker sphincters.[19] It is rarely seen in female patients with vestibular fistula, as most have reasonably good sphincter strength except when associated sacral defect (as seen in one of our case).

All incontinent patients were subjected to intensive "Bowel management programme" which included diet, laxatives, suppository/enemas and saline rectal wash in incremental manner as needed. Rectal wash was required in all cases of recto-vesical fistula and occasionally in recto-prostatic fistula with very satisfactory results. Others needed various combinations of dietary changes, bowel training/behavioural therapy, laxatives, occassional enemas/suppositories with good results. All patients showed significant improvement after this management and reached scores of 5-6 post treatment (all were assessed again on Kelly's scoring). These results are summarised in table 9. Similar management programme was used which showed significant improvement in a study by Iwai et al.[19] Three patients reached no soiling, no incontinence (i.e., no fecal leak) status. Out of this one patient of recto-vesical fistula had an initial score of 1 and now has score of 5, while two other patients reached a score of 6 (both vestibular fistula). This case of recto-vesical fistula with extremely poor score of 1 showed dramatic improvement with rectal wash. Another patient of recto-vesical fistula showed improvement upto score 4 (initial score-3). Three other patients reached score of 5 (with very occasional/rare staining or incontinence). Thus, in our study we could achieve continence in 70.8% patients in post-operative period which improved to 83.3% patients after treatment (Bowel Management), which is a very satisfactory number. The few (four) patients, who still had fecal leak post bowel management, actually had significant improvement with very occasional/rare accidents vis -a- vis their previous condition (Table 9). Our rates were different for recto-bulbar and recto-vestibular fistula and were significantly lower and higher respectively than comparative study. Total number of incontinence cases was lower.[5] As discussed, these cases of vestibular fistula most likely had overflow incontinence due to constipation rather than neuromuscular/ sphincter problem, The satisfactory results found post bowel management shows it is possible to provide them with a tolerable and reasonable/good quality of life. Thus, the majority of these children will engage in typical activities for their age.

First line of treatment for incontinence should be non-surgical. Usually, three categories of these patients are seen-(1) Those with high anomaly, a flat bottom, weak muscles, a sacral abnormality (2) Individuals with good muscle quality and a healthy sacrum but a surgically misplaced bowel with respect to sphincters. (3) People who consistently experience overflow incontinence due to severe constipation with a very dilated recto-sigmoid. Each group needs particular type of management. Patients with poor anatomy require some sort of bowel management programme that makes use of saline rectal wash/enema/suppositories to ensure adequate emptying and clean periods. If that fails, Malone's method, muscle transfer or tightening surgery, biofeedback mechanisms, and other approaches to bowel control and hygiene are available.[22] Long-term outcomes are yet to be seen, although employing this treatment may help avoid permanent colostomies. We neither used nor needed any of above methods in our patients. Patients in 2nd category might be candidates for reoperation to correctly place the bowel, though its outcome varies with only 15-46% of patients achieving worthwhile continence. Third group of patients might be candidates for anterior resection.[5] A permanent end stoma might be best for cases of incontinence where all other therapies have failed.

Our study had 2 rare cases -one "H" type recto-vaginal fistula and second type IV pouch colon. The detailed classification and description of these anomalies will be beyond scope of this study. Hence, we describe them in brief. The first case presented at one month age with swelling and inflammation at introitus followed by faecal discharge. On examination was found to have recto-vaginal fistula. It most likely was a case of acquired fistula following a local abscess. It was repaired as staged procedure with excellent result. The second case was that of type IV pouch colon. In first stage, transaction of fistula (recto-vesical) and complete resection of pouch was done with end colostomy, 2nd stage was done as abdomino-perineal PSARP. Though he had significant incontinence but following aggressive "bowel management" overall result was satisfactory.

Table 9: Fecal leak: relevant clinical details, treatment used and improvement in Kelly's score in patients with fecal leak.

Fecal leak (incontinence + staining)								
Type of anomaly	**Sex**	**Nature of fecal accident (incontinence/ staining)**	**Anal tone/ sphincter**	**Sacral anomaly**	**Kelly's score (initial)**	**Treatment used**	**Current status**	**Post treatment Kelly's score**
Recto-prostatic fistula	M	Occasional incontinence + staining	Weak	2 sacral bodies	3	Rectal wash/ enema bowel training diet	Improvement (very rare accident (staining)	5
Recto-vesical fistula	M	Always incontinent	Weak	2 sacral bodies	1	Rectal wash (needs once a day)	Significant improvement (no staining or incontinence after washes)	5
Pouch colon (type IV)	M	Occasional incontinence + frequent staining	Average /weak	-	4	Enema (occasional rectal wash)	Improvement (occasional staining)	5
Recto-vesical fistula	M	Incontinence (frequent)	Weak	2 sacral bodies	3	Rectal wash	Improvement (occasional incontinence)	4
Vestibular fistula	F	Occasional Incontinence (overflow)	Strong	-	4	Laxatives Regular enema/ suppository diet, behaviour therapy	Improvement (very rare staining)	5
Vestibular fistula	F	Occasional incontinence (overflow)	Average /weak	2 sacral bodies	4	Laxatives Bowel training (behaviour)	Fully cured (continent)	6
Vestibular fistula	F	Occasional staining (overflow)	Strong	-	4	Laxatives (if required) bowel training	Fully cured (continent)	6

PSARP cemented its role as the standard procedure. ASARP was done in two cases presenting late, one aged 6 years and other 16 years, as classical prone "frog" position was not possible due to larger body size. But it achieved equally good results. Post-op anal dilatation is must and done according to age relevant dilator size for 6 months to 1 year. Sacral defect remained an important indicator for assessing future prospects of continence and all cases with sacral defects had incontinence to some extent. We did definitive procedure (PSARP/ASARP) for high ARM around 10 months to 1 year age, though the range was 6 months to 16 years as per the patient's arrival to our OPD. We did not do neonatal PSARP (except for low arm for which anoplasty was done).

Limitations

We gave precedence to safety, feasibility and our own experience and learning curve to decide about the staging and timing of definitive procedure. Our study is somewhat limited by number of cases but despite that succeeds in covering wide spectrum of ARM anomalies.

Newer trends are (1) neonatal PSARP, done in neonatal period as soon as patient is fit for surgery with or without colostomy, (2) early PSARP**,** now being done at 4-8 weeks as mentioned in protocol algorithm, (3) laparoscopic PSARP, where abdomino-perineal route is required while doing definitive pull through (for the abdominal part).

CONCLUSION

PSARP remains the standard and most popular technique. If colostomy has been done, distal colostogram is most important in planning surgical approach and gives an idea

about the prognosis. Other investigations like MCU, IVP, echo-cardiography are used when required. MRI to evaluate tethered cord syndrome (reported to be as high as 25% Pena et al may be done, but is still not a part of standard basic protocol including ours.

The decision to do a primary or staged procedure depends on many factors which include surgeon's expertise, success in complete bowel preparation (which in turn depends on time of presentation and fecal load), age of the child, type of anomaly and its complexity. Perineal fistulas (low ARM) and vestibular fistulas are now routinely performed as primary procedures but we performed staged surgeries in two male patients with complex low ARM (a very anteriorly placed anus with bifid scrotum) and also in many vestibular fistulae (due to large fecal load and unsuccessful bowel preparation). Studies have proven that both give the equally good result.

Scoring systems help in analyzing results and formulate strategies which can significantly improve quality of life as discussed. Both constipation and incontinence have to be aggressively treated. Higher anomalies, major sacral defects and redo surgeries are strongly suggestive of future incontinence/ poorer outcome.

Recommendations

Our study has shown that with meticulous planning, efficient surgery with no compromise in surgical principles (with exact placement of bowel within the muscle sphincter complex) and aggressive and appropriate post-operative bowel management, it is possible to give a low surgical complication rate and a reasonable to very good quality of life in most patients. Our study has highlighted the importance of scoring systems (like Kelly's) for post-operative result analysis, planning management for fecal incontinence and also to test success of these endeavors. Expectations should be realistic for both the doctors and parents/care takers, as in most cases the sphincter muscle development, rectal sensation and defecation reflex are poorer than normal comparable subjects. Use of muscle stimulator is highly recommended. The single most important pre-requisite for Primary PSARP is proper and complete bowel preparation. If this cannot be achieved, staged surgery should be done.

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Ethical approval: The study was approved by the Institutional Ethics Committee

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