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A DESCRIPTIVE STUDY OF CLINICAL PROFILE AND FUNCTIONAL OUTCOMES OF CHILDREN WITH ANORECTAL MALFORMATIONS WHO UNDERWENT SURGERY

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Introduction: Anorectal malformations present with an incidence of 1.0 to 1.5 per 5,000 live births slight male preponderance. These malformations include a broad spectrum of defects, low, intermediate, and high, based on the level of the blind rectal pouch with the pubococcygeal and ischial lines on an invertogram. Imperforate anus without fistula is infrequent, accounting for only about 5% of all anorectal malformations.

Aim: This study is aimed to assess the functional outcome and Quality of life of children diagnosed with ARMs and to correlate both with the type of anomaly.

Materials and Methods: The present prospective study was done in a single unit on patients with an anorectal malformation, who presented to Government General Hospital/Guntur Medical College, Guntur from September 2017 to August 2020. Total 34 patients, of these, 27 were Male, and 7 were Female. All patients underwent a detailed clinical examination, evaluation, and management, which were done in the neonatal period. Method for assessment of outcome described in Krickenbeck 2005 (patient age > 3 years, no therapy) has been used. The analyzed data were compared with other series in literature and discussed. A master chart dealing with all aspects has been designed and presented. Statistical analysis is done with SPSS Version 16 and the Chi-square test.

Results: Total patients of 34 cases were followed up who had undergone procedures for anorectal malformation from 2017 to 2020. The ages were above 3years with a minimum follow-up period of 6 months. Of the total, 27(79.41%) were Males, and 7 (20.58) were Females.

Conclusion: The Associated anomalies were noted. The further management of patients with these malformations can be complex. This frequency of associated anomalies in patients with Anorectal Malformations ranges from 40% to 70%[1]. There is significant morbidity related to fecal incontinence after surgical correction. The effect of this on the child's quality of life has received little attention in the surgical literature previously. The evaluation of functional outcome is hampered by the lack of a universally accepted scoring system for fecal continence

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and variations in classification of the types of anorectal malformation. At the Krickenbeck meeting in 2005, a consensus was achieved concerning the assessment of outcome after Anorectal Malformationsrepair[2]. The last four decades have witnessed huge changes in the management of children with Anorectal Malformations. It seems reasonable to propose that the near-universal adoption of posterior sagittal anorectoplasty (PSARP) as the standard operative approach and the simultaneous widespread application of bowel management strategies have together improved social continence in affected children[3].

Key Words: Anorectal malformations, Quality of life, posterior sagittal anorectoplasty.

INTRODUCTION

Anorectal malformations present with an incidence of 1.0 to 1.5 per 5,000 livebirths slight male preponderance. These malformations include a broad spectrum of defects, low, intermediate, and high, based on the level of the blind rectal pouch with the pubococcygeal and ischial lines on an invertogram. Imperforate anus without fistula is infrequent, accounting for only about 5% of all anorectal malformations.[1]

Their association with various congenital anomaly frequently mentioned in the literature, varying among different studies, between 40 and 60 %. The etiology of such malformations remained unclear and are probably multifactorial. There is a low rate of association in families. However, some seem to have an autosomal dominant inheritance pattern with a high incidence of 1 in 100.[2] About half of the children with ARM have associated anomalies. The anomalies associated with anorectal malformation are variable, but most groups agree to the genitourinary anomalies (40-50%) are most prevalent, followed by cardiovascular (30-35%), spinal cord tethering (25-30%), gastrointestinal anomalies (5-10%), and VACTERL (4-9%) anomalies. The higher the rectal pouch is present, the higher are its chances of being associated with defects. Hence, they are frequently associated with rectovesical fistula.[3]

The results of surgical correction of anorectal malformations are assessed clinically by using various scoring systems, or functionally by using anorectal manometry study, neural electrophysiological studies, magnetic spinal simulation, and fecoflowmetry.[4] A significant proportion of children with anorectal malformations have long term problems with fecal continence. Quality of life has become an essential issue in the medical community as well as in support groups for the chronically ill. Pediatric surgeons working with anorectal malformation have significant responsibilities not only regarding the surgical procedure Itself, as well as following-up with patients.[5]

This prospective study is done on patients of anorectal malformations who presented to the Institute of Government General Hospital / Guntur Medical College, Guntur. This study aimed to determine the long-term outcome of surgical intervention at our institution for ARM from the patient's viewpoint as assessed by a questionnaire.

AIMS AND OBJECTIVES

1. To study and assess the functional outcome, quality of life by using Scores and correlating with the type of anorectal malformation and surgery done.

2. To evaluate the associated anomalies which could influence functional outcomes.

3. To study the epidemiology of anorectal malformation.

4. A demographic survey of the presenting cases.

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MATERIALS AND METHODS

The present prospective study was done in a single unit on patients with an anorectal malformation, who presented for review to Government General Hospital/Guntur Medical College, Guntur from September 2017 to March 2020. Of these, 27 were male, and 7 were female patients. All patients underwent a detailed clinical examination, evaluation, and management, which were done in the neonatal period.

The associated anomalies were noted. The further management of patients with these malformations can be complex. This frequency of associated anomalies in patients with ARMs ranges from 40% to 70%. There is significant morbidity related to fecal incontinence after surgical correction. The effect of this on the child's quality of life has received little attention in the surgical literature previously. The evaluation of functional outcome is hampered by the lack of a universally accepted scoring system for fecal continence and variations in, classification of the types of anorectal malformation. At the Krickenbeck meeting in 2005, a consensus was achieved concerning the assessment of outcome after ARM repair.41 The last four decades have witnessed huge changes in the management of children with ARMs. It seems reasonable to propose that the near-universal adoption of posterior sagittal anorectoplasty (PSARP) as the standard operative approach and the simultaneous widespread application of bowel management strategies have together improved social continence in affected children.

In common with other pediatric surgical centers, the last two decades have led to surgical sub-specialization, the emergence of specialist nursing support, the availability of new investigative modalities, and the development of a dedicated clinic for children with this condition. This study is aimed to assess the functional outcome and Quality of life of children diagnosed with ARMs and to correlate both with the type of anomaly. Method for assessment of outcome described in Krickenbeck 2005 (patient age > 3 years, no therapy) has been used.

The analyzed data were compared with other series in literature and discussed. A master chart dealing with all aspects has been designed and presented. Statistical analysis is done with SPSS VERSION 16 and the Chi-square test.

Inclusion Criteria:

1. Age greater than three years.

2. A patient who is not undergoing therapy for incontinence.

3. Patients who underwent surgery for all types of anorectal malformations in Government General Hospital, Guntur.

Exclusion Criteria:

1. Age less the three years.

2. Children with developmental delays and already on therapy for incontinence.

OBSERVATIONS AND RESULTS

Total patients of 34 cases were followed up who had undergone procedures for anorectal malformation from 2017 to 2020. The ages were above 3years with a minimum follow-up period of 6 months. Of the total, 27 were males, and 7 were females.

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TABLE 1: SEX INCIDENCE

	NUMBER (n)	%
MALE	27	79.41%
FEMALE	7	20.58%

The type of anorectal malformation classified based on kruckenbecks classification included Perineal (20%), Rectouretheral (41%), Rectovesical (8%), Pouch colon (5%), Vestibular (17%) and Cloaca (3%) of the total patients taken up for study.

TABLE 2: TYPE OF ANORECTAL MALFORMATIONS SEEN IN MALES

TYPE OF FISTULA	NUMBER(n)	%
PERINEAL	7	20.58%
RECTOURETHERAL	14	41.17%
RECTOVESICAL	3	8.82%
POUCH COLON	2	5.88%

TABLE 3: TYPE OF ANORECTALMALFORMATION SEEN IN FEMALES

In female patients, the most common anomaly was observed to be vestibular fistula, accounting up to three quarters of the patients followed up (85.71%).

TYPE OF FISTULA	NUMBER(n)	%
VESTIBULAR	6	85.71%
CLOACA	1	14.29%

Associated anomalies were found in 71% of males compared to 29% females. Skeletal anomalies were present in 4(28.57%) patients with three sacral agenesis and one limb anomalies. Genitourinary anomalies were found in 6(42.8%), which included Vesicoureteral reflux, Neurogenic bladder, single kidney, and Undescended testis.

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TABLE 4: ASSOCIATED ANOMALIES

ASSOCIATED ANOMALIES	NUMBER(n)	%
GENITOURINARY	6	42.85%
CARDIAC	3	21.42%
SACRAL AGENESIS	3	21.42%
LIMB	1	7.14%
HYPOTHYROID	1	7.14%

Most of the patients underwent PSARP (47.05%) for the anomaly. Perineal fistula group had anoplasty; Cloaca required abdominoperineal pull through. Vestibular anus had ASARP, and one case had minimal PSARP.

TABLE 5: NATURE OF TREATMENT

TYPE OF SURGERY	NUMBER (n)	%
PSARP	18	52.94%
PERINEAL	7	20.58%
ABOMINO PERINEALPULL THROUGH	4	11.76%
ASARP	5	14.70%

TABLE 6: POSTOPERATIVE COMPLICATIONS

Surgical complications	Number(n)	%
Skin excoriation	5	50%
Wound infection	2	20%
Intestinal obstruction	1	10%
Mucosal prolapse	2	20%

TABLE 7: VOLUNTARY BOWEL FUNCTION

Voluntary bowel function	Number(n=34)	%
yes	27	79.41%

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no	7	20.58%

TABLE 8: SCORING (FOR SOILING)

Soiling	NUMBER (n=34)	%
No soiling	16	47.05%
Grade 1	5	14.70%
Grade 2	6	17.64%
Grade 3	7	20.58%

TABLE 9: SCORING (FOR CONSTIPATION)

CONSTIPATION	**NUMBER (n=34)**	**%**
No	18	52.94%
GRADE 1	3	8.82%
GRADE 2	7	20.58%
GRADE 3	6	17.64%

Table 10: Comparison of Type of Anomaly and Voluntary BowelMovement

Type of anorectal malformation and voluntary bowel movement shows a significant association.

TYPE OF FISTULA	**VOLUNTARY BOWEL MOVEMENT**		**p-value**
	YES	**NO**	
PERINEAL	6(85.71%)	1(14.28%)	0.008
RECTOVESICAL	1(25%)	3(75%)	
RECTOURETHRAL	13(92.85%)	1(7.14%)	
CLOACA	0	1(100%)	
VESTIBULAR	6(100%)	0	
POUCH COLON	0	2(100%)	

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TABLE 11: COMPARISON OF TYPE OF ANOMALY AND OUTCOME SCORING FOR SOILING (p-value: 0.91)

No statistical association is seen in this relationship.

TYPE OF FISTULA	SOILING			
	NO	GRADE 1	GRADE 2	GRADE 3
RECTOURETHERAL	7(50%)	3(21.42%)	3(21.42%)	1(7.14%)
PERINEAL	6(85.71%)	0	0	1(14.28%)
RECTOVESICAL	0	0	1(25%)	3(75%)
POUCH COLON	0	0	0	2(100%)
VESTIBULAR	3(50%)	2(33.33%)	1(16.66%)	0
CLOACA	0	0	0	1(100%)

TABLE 12: COMPARISON OF TYPE OF ANOMALY AND OUTCOME SCORINGFOR CONSTIPATION (p-value: 0.229)

Constipation and type of malformation relationships aren't statistically significant.

TYPE OF FISTULA	CONSTIPATION			
	NO	GRADE 1	GRADE 2	GRADE 3
RECTOURETHERAL	6(42.85%)	2(14.28%)	3(21.42%)	3(21.42%)
PERINEAL	2(28.57%)	0	3(42.85%)	2(28.57%)
RECTOVESICAL	4(100%)	0	0	0
POUCH COLON	2(100%)	0	0	0
VESTIBULAR	3(50%)	1(16.66%)	1(16.66%)	1(16.66%)
CLOACA	1(100%)	0	0	0

TABLE 13: COMPARISON OF TYPE OF SURGERY AND VOLUNTARY BOWELMOVEMENT

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Good bowel control is seen after PSARP, Perineal approach than abdominoperineal pull through

TYPES OF SURGERY	VOLUNTARY BOWEL MOVEMENTS	
	YES	N0
PSARP	15(83.33%)	3(16.66%)
PERINEAL	6(85.71%)	1(14.28%)
ABDOMINO PERINEAL PULL THROUGH	1(25%)	3(75%)
ASARP	5(100%)	0

TABLE 14: COMPARISON OF TYPE OF SURGERY AND OUTCOME SCORING FOR SOILING

Soiling is seen more commonly in cases with abdominoperineal pull-throughcompared with the perineal and anterior sagittal approach.

TYPE OF SURGERY	SOILING			
	NO	GRADE 1	GRADE 2	GRADE 3
PSARP	7(38.88%)	4(22.22%)	4(22.22%)	3(16.66%)
PERINEAL	6(85.71%)	0	0	1(14.28%)
ABDOMINO PERINEAL PULLTHROUGH	0	0	1(25%)	3(75%)
ASARP	3(60%)	1(20%)	1(20%)	0

TABLE 15: COMPARISON OF TYPE OF SURGERY AND OUTCOME SCORING FOR CONSTIPATION

In comparison with the type of surgery and different grade of constipation, Constipation is observed in most of the cases with the perineal procedure and posterior sagittal anorectopexy.

TYPE OF SURGERY	CONSTIPATION			
	NO	GRADE 1	GRADE 2	GRADE 3
PSARP	9(50%)	2(11.11%)	3(16.66%)	4(22.22%)
PERINEAL	2(28.57%)	0	3(42.85%)	2(28.57%)
ABDOMINO PERINEAL PULL THROUGH	4(100%)	0	0	0
ASARP	3(60%)	1(20%)	1(20%)	0

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TABLE 16: Association between Associated anomalies and voluntary bowel movements after definitive surgery

On statistical analysis, the p valve was found to be significant, when the relationship of the associated anomaly and voluntary bowel movement is taken.

Associated anomalies	VBM		p-value
	No	Yes	
Bilateral hydronephrosis	1	0	
Bilateral hydronephrosis and undescended testis	1	0	
Cardiac	0	3	
Hypothyroid	0	1	0.002
Radial anomaly	0	1	
Sacral agenesis	2	0	
Sacral agenesis,hypospadias	1	0	
Vesiculoureteric reflex	0	2	
Vesiculoureteric reflex and Neurogenic bladder	1	0	
Renal agenesis	0	1	
No anomaly	1	19	
Total	7	27	

TABLE 17: ASSOCIATION BETWEEN ASSOCIATED ANOMALIES AND SOILING AFTER DEFINITIVE SURGERY

There is no statistical significance of associated anomalies with soiling

Associated anomalies	SOILING		p-value
	No	Yes	
Bilateral hydronephrosis	0	1	
Bilateral hydronephrosis and undescended testis	0	1	
Cardiac	1	2	
Hypothyroid	1	0	0.436
Radial anomaly	1	0	
Sacral agenesis	0	2	
Sacral agenesis,hypospadias	0	1	
Vesiculoureteric reflex	1	1	
Vesiculoureteric reflex andNeurogenic bladder	0	1	
Renal agenesis	0	1	
No anomaly	12	8	
Total	16	18	

TABLE 18: ASSOCIATION BETWEEN ASSOCIATED ANOMALIES AND CONSTIPATION AFTER DEFINITIVE SURGERY

Associated anomalies	CONSTIPATION		p value
	No	Yes	
Bilateral hydronephrosis	1	0	

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Bilateral hydronephrosis and undescended testis	1	0	0.277
Cardiac	0	3	
Hypothyroid	0	1	
Radial anomaly	0	1	
Sacral agenesis	2	0	
Sacral agenesis,hypospadias	1	0	
Vesiculoureteric reflex	1	1	
Vesiculoureteric reflex andNeurogenic bladder	1	0	
Renal agenesis	0	1	
No anomaly	11	9	
Total	18	16	

DISCUSSION

This study is done in children who had definitive surgery for anorectal malformation and followed and graded with a Krickenbeck score[2]. A total of 34 children were included in the study. Children were followed up over the course of the study from the time of colostomy closure or definitive procedure. The minimum time of follow up was six months, and the age of patients ranged from 4 years to 14 years by the time study got completed. The majority are male 27(79.41%) and it might represent a true male preponderanceor maybe because many girls are not brought for follow up in our society[4]. As Surgical techniques evolved, different anorectal malformation classification and different scoring systems for assessing bowel function in patients were introduced. The latest classification Krickenbeck, which identified eight major types of anorectal malformations and rare/regional variants[2].

In our study, thirty-four children met the inclusion criteria having more than three years of age and no developmental delays by the end of the study period. There is a male preponderance of males as observed in different study groups[4], As observed by Brisighelli G et al., Makrufardi F et al., and Swaleh S et al[5,6,7], which was around 52%, 69.8%, and 56%. This is a general trend observed all over the world, with men more affected by this anomaly[4]. In our study, the most common type of fistula observed is Rectourethral (41.17%) seen in males[8]. Concerning the types of fistula, Makrufardi F et al. and Swaleh S et al., observed 25.5% and 32% cases with rectourethral who responded to the questionnaire[6,7]. The type of anorectal malformation classified based on Kruckenbeck classification included Perineal (20%), Rectouretheral (41%), Rectovesical (8%), Pouch colon(5%),Vestibular(17%) and Cloaca (3%). This helps to further analyse the data in a relevant manner, and appropriate scoring can be used. Perineal and Vestibular anomalies are the low anomalies accounting 37% of patients in the group. They underwent a Perineal, Anterior sagittal approach, and minimal PSARP as required. Rectourethral had seven cases, each of the bulbar and prostatic fistula in our study. Most of them underwent PSARP with colostomy as a staged procedure.

One of the regional variants seen was the Pouch colon. A high incidence of pouch colon is reported from the Indian subcontinent up to 40%as per classification by Rao et al[9]. In our study there are two such cases that were followed up.Standard procedure done was a three-staged procedure in both cases.

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in our study, Associated anomalies were 41.17% which are similar to what is reported by Endo M et al[10].Associated anomalies were found in 71% of males compared to 29% females. Cho S et al. also reported a similarly high incidence of associated anomalies In males[11].They also reported high anorectal malformations in males compared to females, similar to our study. Skeletal anomalies were present in 4(28.57%) patients with three sacral agenesis and one limb anomalies. Radial bone anomaly is seen in one patient.

Genitourinary anomalies were found in 6(42.8%), which included 3 cases of Vesicoureteral reflux, two cases of bilateral hydronephrosis. One patient had neurogenic bladder with vesicoureteric reflux, renal agenesis is seen in two cases, one case of the undescended testis with bilateral hydronephrosis and one case was associated with penoscrotal hypospadias. Two cases of congenital pouch colon had associated genitourinary anomalies, one had neurogenic bladder with vesicoureteric reflex and another had bilateral hydronephrosis with left undescended testis The Genitourinary system was the most affected organ system, as seen in other studies[8,12].·Therefore, in any case of anorectal malformation, it is imperative to examine the genitourinary system. Ultrasound, MCUG plays an important role in the follow up of the patient[12,13]. Spinal agenesis was evaluated with ultrasound and MRI, Cardiovascular anomalies (21.42%) are seen in three patients, with septal defects being the most common, as patients, severe abnormalities usually don't survive. Screening echocardiogram is done in all patients at birth and before definitive surgery.Cardiac anomalies (38%) were reported to be most common by Qazi et al[14].

The posterior sagittal approach was first introduced by Pena[15], which is now the gold standard worldwide. Most of the patients underwent PSARP (52.94%) for anorectal malformation, especially rectourethral fistula. Perineal approaching useful in seven (20.58%) patients. Others required Abdominoperineal pull through (11.76%) and anterior sagittal approach (14.70%). Surgical complications are seen in 29% of patients in the immediate postoperative period. These include skin excoriation, wound infection is seen in two cases of abdominoperineal pull through. Intestinal obstruction was seen in one case after colostomy closure.

Evolution similar to that of classification of anorectal malformation was seen in scoring systems also. Scott, in 1960 was the first person to propose a scoring system. This was based on defaecation habits, stool control, perianal soreness, and the function of puborectalis on per rectal examination. Later Kelly, Holschneider, Pena, and Rintala proposed different scoring systems over the years. Finally, the Krickenbecks scoring system is developed with consensus in 2005[2,15]

There is a lot of change after the introduction of PSARP and classification by Pena[3,15]. The requirement of continuing care of the patients after the definitive procedure is discussed. There is a need for frequent clinical reviews, proper education of parents, and the patient. The requirement of a single scoring system with a standardized classification of the anorectal anomaly and surgical procedure category would help in streamlining treatment and followup, thereby improving the quality of care. These special children require the involvement of multiple professionals from different fields. Our study scope is limited by the number of patients, period of study, and variations of responses by mothers, especially in grading and their perceived interpretation. Newer modalities such as MRI can play an important role in the case that can be redone based on the anatomical changes after the initial surgery. It can decide

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the plan for redo surgery if it can help. Low anorectal malformations have a good functional outcome, require to follow up avoiding long term complications.

CONCLUSION

The incidence of Anorectal malformations in males appears to be higher than females Rectouretheral fistula is the most common anomaly in males and Vestibular fistula is the most common presentation of in females. The most common Associated anomalies in this study are genitourinary anomalies. Most frequent is the Vesicoureteric reflux seen in three patients. The incidence of Associated anomalies is higher in patients with high Anorectal malformations. Both cases of Congenital Pouch colon of type III had associated genitourinary anomalies.Sacral agenesis observed in three patients, each with a different type of fistula.

PSARP is the most common surgical procedure performed.Skin excoriation due to soiling is the most common complication in the immediate postoperative period.Most of the patient had voluntary bowel control after Surgery. High anorectal malformations and cases with Sacral agenesis observed to lose voluntary bowel control. Low Anorectal malformations, including perineal and vestibular fistula, the most common complication is constipation. Cases with Pouch colon and Sacral agenesis suffered severe soiling. Both cases of Pouch colon underwent the Abdominoperineal pullthrough procedure, had a loss of voluntary bowel control, developed fecal and urinary incontinence. Most of the cases of Perineal and Vestibular fistula had good fecal continence, did well on follow up.Rectovesical fistula is associated with soiling in most of the cases. Patient with Cloaca is associated with fecal incontinence and soiling with loss of voluntary bowel control. Patients with Abdominoperineal pull-through had a higher incidence of soiling and fecal incontinence. Standardization of type of fistula, type of Surgery, and follow up the scoring system are required to improve the outcome of the patients. Newer modalities like MRI can give excellent soft-tissue characterization, multiplanar imaging, and lack of ionizing radiation.

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