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An insight into the demographic profile and outcomes of Anorectal Malformations from a resource limited tertiary care institute in Western India

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Abstract

Aim: Anorectal anomalies in neonates are discussed with a focus on imaging for diagnosis, categorization, and assessment of severity before surgical therapy is provided. The study's primary objective was to examine the demographics and results of Anorectal Malformations from a resource-limited tertiary care facility in Western India.

Subjects and Methods: This was a prospective observational study conducted at the Department of Paediatric Surgery, SMS Medical College & SPINPH Jaipur which included 230 babies with ARM who were admitted during last one year. All included patients were evaluated with detailed clinical history and examination for the diagnosis and the type of ARM, associated congenital anomalies and outcomes after surgery.

Results: In this study, there were 230 neonates. Of these, 173 were male and 57 were females (M: F = 3:1). High-type ARM was seen in 192 (83.47%) patients, while low-type ARM in 38 (16.52%) neonates. Associated

malformations were present in 38.26% (n=88) neonates with higher incidence in HARM (42.18% vs 18.42%). CPC was found in 36 patients. The average birth weight was 2420 grams (range 930 to 3500 grams). Associated anomalies were identified in 88 (38.26%) patients, with gastrointestinal anomalies being the most common (37.5%). There were two patients with EA, DA with ARM, 26 patients with EA with ARM and one patient was having DA with ARM.

Conclusions: We conclude that a greater mortality rate was associated with the neonates having a low birth weight, double or triple atresia, neonatal GIT perforation, infection upon arrival, and esophageal and cardiac defects. A multifaceted strategy that emphasises early resuscitation, rapid referral, stringent infection control, optimal feeding, perfect physiologic state and availability of bedside echocardiography and ultrasonography is required to reduce infant death and morbidity.

Keywords: Anorectal mal formation, associated anomalies, colostomy, neo natal mortality.

Introduction

Anorectal mal formations (ARM) are diverse and are among the more frequent congenital anomalies encountered in paediatric surgery, involving the lower gastro intestinal tract, urinary and /or genital system [1].

The anus and rectum, as well as the urinary and genital systems, are all involved in anorectal anomalies, which may affect both boys and girls. Its prevalence is estimated at one per 5,000 live births. Anorectal malformations range from the least complex and most easily cured to those that are more complex, difficult to manage, and often accompanied by other anomalies, affecting severity and functional prognosis. Surgical techniques for correcting these anomalies underwent a significant shift in 1980 with the development of the posterior sagittal approach, which gave surgeons a better view of the recto-genitourinary junction anatomy and enabled them to perform repairs under direct vision [2]. Advances in imaging technology and a more complete understanding of the pelvic structures' architecture and physiology at birth have led to a more accurate diagnosis, as well as a more accurate prognosis. The surgeon's primary objectives are bowel control, urine control, and sexual function when repairing these defects [3]. The greatest opportunity for a functional result may be achieved with early identification, proper care of related abnormalities, and precise surgical repair. Even after a satisfactory anatomic repair, faecal and urine incontinence might arise because of related issues such a poorly formed sacrum, inadequate nerve supply, and spinal cord malformations [4]. These patients' quality of life has been improved by a successful bowel management regimen that includes enema and food modifications. Prognosis and quality of life of the patient is determined by holistic management. Various risk factors like anorectal malformation type, associated congenital anomalies, sepsis, and prematurity are associated with morbidity and mortality in these patients [5]. Anorectal malformations may be affected by delays in diagnosis, referral, and the existence of related defects (ARM). It was the goal of this research to examine the early postoperative outcomes of newborns with ARM [6]. This observational study was conducted to understand the demography and various types of anorectal malformations along with associated malformations and their outcomes.

Materials & methods

This was a prospective observational study conducted in the Department of Paediatric surgery, SMS Medical College and attached SPINPH, as a part of registry for common surgical conditions during the period from January 2021 to December 2021. After ethical clearance

(306/MC/EC/2021) all neonates with ARM, admitted during this period were included in the study. Patients who presented beyond neonatal period or who underwent any surgical intervention outside our hospital were excluded from the study.

All included patients were evaluated with detailed clinical history and examination for the diagnosis of the type of ARM, and associated congenital anomalies. Written informed consent was taken for necessary procedures.

Radiological examination (Babygram and X-ray prone cross table lateral view with raised pelvis- (after 18 to 24 hours of life) was done in neonates without visible fistula. Level of distal gas shadow; findings of presence of intestinal atresia, congenital pouch colon (CPC), associated anomalies and bowel perforation were noted.

Abdominal ultrasound to detect any other abdominal pathology and 2D Echocardiography in patients suspected to have cardiac anomalies were done.

Preoperative resuscitation was done with intravenous fluids, nasogastric suction and broad-spectrum antibiotics along with measures to prevent hypothermia and hypo glycaemia. When required, respiratory support was given. These were continued in post-operative period also. Patients were kept nil orally till the stoma started functioning or the child had passed meconium.

In neonates with oesophageal atresia, oral feeds were attempted only after ano esophagogram was done on the 5^{th} postoperative day to detect anastomotic leaks. Total parenteral nutrition was provided where oral feed was restricted for more than five days.

Surgical Interventions For anorectal mal formations

All patients with low type of defects underwent primary anoplasty without a protective diverting colostomy while patients with high- type ARM underwent a diverting colostomy. Colostomies were either left transverse or high sigmoid colostomy. Depending on the type of CPC, the neonates underwent fistula ligation with a) pouch ostomy (Type 1 and Type 2) or b) excision of pouch with end colostomy/ end ileostomy (in rest). In neonates undergoing diverting colostomy, stoma care was explained to the mother along with distal loop wash during the postoperative period (before discharging the patient from the hospital) and during follow-up visits to prevent faecaloma formation. In the present study of early outcome in neonates with ARM, data was analysed and their impact on neonatal mortality and morbidity was studied.

Statistical analysis

Data were coded and recorded in MS Excel spreadsheet program. SPSS v23 (IBM Corp.) was used for data analysis. Descriptive statistics were elaborated in the form of means ± standard deviations and medians or IQRs for continuous variables, and frequencies and percentages for categorical variables. Group comparisons for continuously distributed data were made using independent sample‘t’ test when comparing two groups. If data were found to be non-normally distributed, appropriate non-parametric tests in the form of Wilcoxon Test were used. Chi-squared test was used for group comparisons for categorical data. Statistical significance was determined at P value < 0.05.

Results

Table 1: Type of anomalies in the present study.

Male	N (%)	Female	N (%)
Perineal fistula	36 (15)	Perineal fistula	2 (3.5)
Recto-urethra l/ bladder neck fistula or without fistula	133 (57.82)	Vestibular fistula	20 (8.69)
		Persistent cloaca	15 (26.31)
		Imperforate anus without fistula	18 (31.57)
Rectal atresia	4 (1.7)	Rectal atresia	2 (3.5)
Total	173 (75)		57 (25)

Table 2: Congenital Pouch Colon and its outcome

Type	Frequency	Survival (%)	Mortality (%)
CPC 1	5	4 (80)	1 (20)
CPC 2	10	8 (80)	2 (20)
CPC 3	3	2 (66.67)	1 (33.33)
CPC 4	18	12 (66.67)	6 (33.33)
Total	36 (15.2%)	26 (72.22%)	10 (27.77%)

Table 3: Percentage of associated mal formations and gender distribution.

Gender	Frequency	Associated Malformations (%)
Male	173	67 (38.72)
Female	57	21(36.84)
Total	230	88 (38.26)

Table 4: Summary of associated anomalies in the present study and their outcome.

Associated Anomalies		Frequency (%) (n=88)	Survivors (%)	Mortality (%)
Gastrointestinal Anomalies	EA	28 (84.84)	5 (17.85)	23 (82.14)
	Duodenal Atresia	3 (9.09)	0	3 (100)
	Meckel's Diverticulum	2 (6.06)	1 (50)	1 (50)
Total		33 (37.5)	6 (18.18)	27 (81.81)
Cardiac Anomalies		19 (21.59)	4 (21.05)	15 (78.9)
Urogenital Anomalies		21 (23.86)	10 (47.61)	11 (52.38)

	Distal Hypospadias	6	-	-
	Proximal Hypospadias	2	-	-
	Undescended Testis	4	-	-
	Scrotal Transposition	4	-	-
	Hydronephrosis	2	-	-
	Posterior urethral valve	1	-	-
	Vaginal Atresia	1	-	-
	Disorders of sex development	1	-	-
Skeletal	Limb Anomalies	6	6	0
	Cleft Palate	4	4	0
Total		10 (11.36)	10	0
Down's syndrome		1 (1.1)	0	1
Inguinal Hernia		4 (4.54)	2	2

Table 5: Double/ Triple at resias and their outcome (out of 88 patients with associated anomalies)

Type	Frequency	Mortality (%)	Survival (%)
EA+DA+ARM	2 (6.8)	2 (100)	0
EA+ARM	26 (89.65)	21 (80.76)	5 (19.23)
DA+ARM	1 (3.4)	1 (100)	0
Total	29 (32.95)	24 (82.75)	5 (17.24)

EA – Oesophageal atresia, DA- Duodenal atresia, ARM- Anorectal malformation.

Table 6: Cloaca and their outcome

Cloaca	Frequency (%)	Discharge (%)	Death (%)
Cloaca	8 (53.33)	6 (75)	2 (25)
Cloaca + Hydro metro col pos / Vaginal Atresia	1 (6.67)	1 (100)	0
Cloaca + CPC 1	3 (20)	2 (66.67)	1 (33.33)
Cloaca + CPC 2	3 (20)	3 (100)	0
Total	15 (6.5)	12 (80)	3 (20)

CPC – Congenital pouch colon.

During the research period, 230 newborns with ARM were admitted. There were 173 male and 57 female newborns (M: F = 3:1). There were 192 (83.47 %) neonates with high-type ARM and 38 (16.52 %) infants with low-type anomaly. In the male category (n=173; 75%), the number of patients with Recto-urethral/bladder neck fistula or without fistula was 133 (57.82%), the number of patients with perineal fistula was 36 (15%), and Rectal atresia was in 4 (1.7%) patients. In the female group (n=57; 25%), vestibular fistula was the most common finding (n=20; 8.69 %), followed by ARM without fistula in 18 patients, Persistent cloaca in 15,

rectal atresia in 2 and perineal fistula in 2 neonates (Table 1). The average weight was 2420 grams (range 930 to 3500 grams). In 36 (15.7%) patients with congenital pouch colon (CPC), type 4 was the most common (n=18; 50%) out of which more than two thirds of patients were discharged home (n=12; 50%). Type 2 was the next common finding (n=10; 27.77%), followed by type 1 and type 3. Among all the neonates with pouch colon nearly two third of these babies were survived (n=26; 72.22%) with the maximum deaths in the type 3 variety (n=1/3; 33.33%) (Table 2).

Associated anomalies were seen in 38.26 % (n=88) of new born, with a higher prevalence in boys (38.72 % vs. 36.84 %) and HARM (42.18 % vs 18.42 %). (Table 3).

Among the associated congenital anomalies gastro intestinal anomalies were the most common association (n=33; 37.5 %). As an individual association with ARM in our series, there were 28 (31.21 %) cases of Oesophageal atresia (EA), three cases of duodenal atresia and two patients had Meckel's diverticulum. Among these 33 patients with gastro intestinal anomalies only 6 (18.18%) patients were discharged and 27 patients died. Associated cardiac defects were noted in 19 (21.6%) patients out of which only 4 (21.05%) patients were survived. Urogenital anomalies were found in 21 (23.86%) patients including hypospadias (n=8), undescended testes (n=4), hydronephrosis (n=2), posterior urethral valve, vaginal atresia and disorders of sex development (n=1). Associated skeletal anomalies noted in 10 patients which included dysplasias, osteoporosis, absent or deformed bones, delayed bone development and limb reduction abnormalities. Cleft palate was noted in four patients. We had one patient with Down's syndrome which could not be survived. Inguinal hernia was observed in four patients (Table 4).

There were 29 (33%) patients with multiple at resias out of which 26 had esophageal atresia and anorectal atresia, two patients had triple atresia (duodenal, esophageal and anorectal atresia) and one patient had duodenal atresia with anorectal atresia. Out of these 29 neonates only five (17.24%) could be survived and rest of the babies succumbed to death in view of multiple congenital anomalies (Table 5).

Cloacal anomalies were seen in 15 (6.5%) patients and cloaca with vaginal atresia, cloaca with CPC type 1, cloaca with CPC type 2 were seen in one patient each (Table 6).

Discussion

Anorectal mal formations occur in 1 in 5000 live births and are among the wide range of congenital mal formations that are often seen in paediatric surgery. In a single centre with constrained resources, our research included a large number (230) of ARM patients.

In contrast to male infants, who often develop abdominal distension over a few days in most patients with a recto urethral fistula, delayed presentation is seen in female children because there is still some passage of meconium/faecal matter via the vestibular fistula. The risk of death for neonates with low birth weight (LBW) is still much greater than for newborns with normal birth weight. About 50 to 60 percent of patients with ARM may have associated abnormalities [1].

As reported by Mathew et al, we also had a high number (n=192; 83.47 %) of high ARM which is in contrast to Goossens et al where they have reported low type malformations in more than 50% of patients [7]. In males we have found that the high ARMs (rectourethral fistula) were present in 57.82% of the patients. Similar findings were also reported by Mathew et al and Levitt et al [1, 2]. These neonates present with either meconuria or flat perineum with abdominal distension. These were

managed by left transverse colostomy in the neonatal period and later on PSARP was done.

We have observed vestibular fistula as the most common (n=20; 8.69 %) defect in female newborns as per the reports of most of the large studies [1, 2]. Babies with vestibular fistula were observed for 24 to 48 hours for decompression of the rectum via fistula. If they did not decompress well and progressive abdominal distension ensues or associated severe congenital anomalies, then a left transverse colostomy was performed. If they decompress well, these neonates were kept on rectal washes and laxatives and definitive procedure was done later on after 3-4 months of age.

Rectal atresia is a rare condition characterised by stenosis or atresia of the rectum with normal anal opening. In our study we had seen rectal atresia in six (2.6%) patients, including four males and two females. They were managed by colostomy in the neonatal period followed by recto-rectal anastomosis later on.

Cloacal anomalies are the fusion of rectum, urinary bladder and vagina to give rise to a common pouch with a single drainage channel. The definitive management of which depends on the length of the common channel. Many of these neonates present with a Hydro metro col pos due to obstruction of vaginal outlet and pooling of urine which leads to dilatation of vagina by fluid palpable per abdomen. They are managed by tube vagin ostomy along with transverse colostomy [1]. Though rare, we have observed cloaca in 15 (6.5%) patients and one patient had Hydro metro col pos. This was the third most common malformation in females after vestibular fistula and ARM without fistula. Six patients (n=6/15; 40%) with persistent cloaca had associated congenital pouch colon (Type 1 & 2; 3 each). The survival among the neonates with cloaca was 80% (12/15), with a mortality of 20% (3/15). The mortality of these three neonates could be attributed to the low birth weight (n=3), delayed referral (n=2), sepsis (n=3) and associated complex congenital anomalies (n=1). Early diagnosis and preoperative evaluation of urogenital, cardiac and spinal anomalies is very crucial in the survival of these babies.

Congenital pouch colon (CPC) is rare and regional variant of ARMs as per Kricken beck classification with an incidence of 2 - 18% in overall cases of ARM. Being a regional variant of Indian subcontinent, this variety is seen in nearly 30-40% cases of ARM in India. In this variety the colon is replace by a pouch like dilatation in its whole or part of the length and the terminal end opens into the bladder or vagina by a narrow fistula [8]. In our series the incidence was 15.2% (n=36) which is similar to the reports by Indian authors [1, 8]. Out of these nearly two-thirds of the neonates were survived and the mortality was 27.7% (n=10). The probable reasons for the high death rate are low birth weight (n=5), delayed presentation (n=2), associated complex anomalies (n=3). As per the classification of CPC there are four types of pouch colon [8]. We have seen Type 4 in majority of patients (n=18; 50%), followed by type 2 (n=10; 27.77%), type 1 (n=5; 13.8%) and type 3 (n=3; 8.3%). A type 5 variety of pouch colon has also been described by Saxena et al, however, we have not observed this variety in our cohort [9].

The type of anorectal malformation and associated anomalies have an impact on the management and outcomes. Levitt and Pena have reported associated anomalies in up to 50% of the patients with ARM [1]. We have also observed associated anomalies in nearly 40% of the patients. in accordance with the literature as compared to neonates with low ARM, those with high ARM are more likely (42.18 % vs 18.42 %) to have related abnormalities [2]. In our series gastrointestinal anomalies were the highest (n=33; 14.3%) in number

which included esophageal atresia (n=28), duodenal atresia (n=3) and Meckel's diverticulum (n=2). This is somewhat lesser than reported (27%) in previous series [10]. The next most common associated anomalies were urogenital anomalies, which is seen in 21 (9.1%) patients. These included hypospadias (n=8), undescended testes (n=4), hydronephrosis (n=2), posterior urethral valve (n=1), vaginal atresia and disorders of sex development (one each). In the western literature urogenital anomalies were the most common type of association reported with the VUR (vesicoureteric reflux) in nearly one thirds of the patients [11].

However, in our series gastrointestinal anomalies were the most common association. We have observed congenital cardiac defects in 19 (8.26%) patients which is lower than the reports by Kamal JS et al, probably due to the non-availability of the echocardiography in the immediate neonatal period [12].

In low type of anomalies standard posterior sagittal anorectoplasty is the preferred surgical option and in high type the diverting colostomy followed by PSARP later on is the choice of surgical technique [10]. However, the management of rare variants like cloaca, pouch colon, rectal atresia and vaginal atresia may require some additional procedures according to the type of the malformations [10]. The management of pouch colon requires careful examination of intra-op findings and decision to do a three-stage procedure (ileostomy, colorrhaphy and pull through, stoma closure) [9].

We have observed a higher neonatal mortality (71/230; 30.9%) in our cohort than reported (6%) in the literature [10]. Highest death rate was observed in the neonates having multiple at resias (24/29; 82.7%) and with the cardiac defects (15/19; 78.94%). Possible explanation for this very high mortality at a tertiary care institute is that ours is the only government institute catering most of the neonatal emergencies in the state and have a very high case load (around 10-15 neonatal emergency per day). This compromises the health facilities and working of the neonatal intensive care staff. Second reason is the poor management of low-birth-weight babies due to unavailability of the neonatology facilities round the clock. Other reasons are late presentations due to delayed referral, poor transport system and sepsis at the time of presentation due to poor hygiene and cord care practices. All these factors contribute to the neonatal deaths even in the tertiary care centre.

During the follow up period constipation, pseudo incontinence, and faecal incontinence are common issued reported from these patients, and they can have a negative impact on their quality of life. The examination of colon and anal sphincters, along with any associated malformations is very crucial for a customised bowel management regimen.

Using both anorectal manometry and endo-anal ultra-sonography, anal sphincters may be studied in detail [13]. The emergence of high-resolution and high-definition anorectal manometry, as well as the three-dimensional reconstruction of endo Sono graphic pictures, has greatly improved the accuracy of displaying pressures and muscle lesions of anal sphincters. Sphincter injuries of varying severity have been described in literature on ARM-affected children, including changes in rest and squeeze pressures and lack of a recto-anal inhibitory reflex (RAI). Because of the clinical diversity of these individuals, the limited series analysed, and the many diagnostic techniques employed, the data acquired are not homogenous [14]. However, patients with ARMs should have a comprehensive manometric and endosonographic examinations to better understand the underlying pathophysiology of their anorectal diseases and guide further therapy.

Conclusion

Anorectal abnormalities being a common pediatric surgical condition needs particular attention towards care in the neonatal period. A greater mortality rate was associated with the neonates having a low birth weight, double or triple atresia, neonatal GIT perforation, infection upon arrival, and esophageal and cardiac defects. A multifaceted strategy that emphasises early resuscitation, rapid referral, stringent infection control, optimal feeding, perfect physiologic state and availability of bedside echocardiography and ultrasonography is required to reduce infant death and morbidity. In order to deliver the treatment with the highest possible standards for infants, a multidisciplinary approach that including neonatologists, paediatric surgeons, anaesthesiologists, and radiologists would be a great asset.

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