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Original research article

A prospective study on clinico-etiological profile of ano-rectal malformations in a tertiary care centre

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Abstract

Background: Imperforate anus or ano-rectal malformations are one the most challenging congenital disorders to manage owing to the complex anatomical structures and young age of patients (neonates). This study aims to assess the clinico-etiological profile of patients presenting to the tertiary care hospital.
Materials and methods: 100 patients with ano-rectal malformations were included in this prospective study which was conducted over 1 year duration in the Department of Pediatric surgery, Niloufer hospital, Hyderabad.
Results: Males were more common than females. High type of ano-rectal malformations was the most common type. Low level of ARM was more common in females. Cardiac anomalies were the most common associated anomalies followed by urological. Amongst urological anomalies, renal agenesis was the most common abnormality detected on ultrasound examination, while VUR was the most common abnormality detected on VCUG.
Conclusion: Anorectal malformations have a male preponderance with associated malformations of other systems. In-utero detection of ARM is challenging and usually these defects are detected after birth. Screening all babies for such malformations and managing them can alleviate significant morbidity and mortality.
Keywords: Anorectal malformation, urogenital abnormalities, high level, vesico-ureteric reflux, posterior urethral valves

Introduction

Anorectal malformations (ARM) are characterized by absence of anal opening (imperforate anus) and the presence of a fistulous tract with one end of the tract opening into the adjacent structures (such as urinary tract in males and internal genital organs in females or anterior to the usual anal opening). The incidence of ARM is 1 in 5000 live births making it one of the most common congenital anomalies [1, 2].
ARM can be broadly classified into two types-"high type" of defects and "low type" of defects. Amussat was the first one to perform the first of its kind anoplasty in the 19th century by suturing the edges of rectal wall to the skin. The low type of defects was managed by perineal surgeries and the high type of defects was being managed by colostomies, which led many of the patients incontinent. With the advent of posterior sagittal approach, surgeons were able to operate easily as this approach offered a direct vision of the complex anatomical structures [3-6].
Anorectal malformations can be associated with malformations of other systems. Sacrum is the most common bony structure to be involved with tethered cord syndrome being the most common sacro-spinal anomaly to be associated with ARM [7, 8]. Associated anomalies of the uro-genital system are seen in approximately 50% of the neonates [3]. Down's syndrome, Townes-Broks syndrome (imperforate anus, dysplastic ears and thumb malformations) [9], Currarino's syndrome (sacral agenesis, imperforate anus and pre-sacral mass) [10] and VACTERL syndrome (vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb anomalies) are few of the syndromes associated with imperforate anus.
The incidence of ARM in Southern India is not well known. Hence the study was taken up to assess the incidence, types, age of presentation, associated anomalies and prognosis after management in patients presenting to our tertiary care center.

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Methodology

This prospective study was conducted in the Department of Pediatric surgery, Niloufer hospital Hyderabad, over 1 year period, i.e., from March 2022 to February 2023. All patients with ano-rectal malformations were included in the study after taking a written informed consent from the parents of the patients.

A detailed history including the demographics, birth history, antenatal history and family history was taken. All patients were subjected to a thorough clinical and systemic examination to identify any associated anomalies of other systems. Radiological investigations included babygram, invertogram, X-ray of lumbosacral spine, ultrasound examination of the abdomen and voiding cysto-urethrogram (VCUG). Anorectal malformations were classified into high, intermediate and low types based on Wingsfield classification.

Ethical committee approval was taken prior to the start of the study. all data was entered and analyzed using Microsoft excel.

Results

110 patients with ano-rectal malformations were included in this study. There were 70 males and 40 females (63.6% and 37% respectively).

Most of the patients presented within first 1 week of birth (94.5%). The remaining 6.5% of patients had presented after 1 week of birth. Low type of defects were common amongst the ones who presented after 1 week of birth.

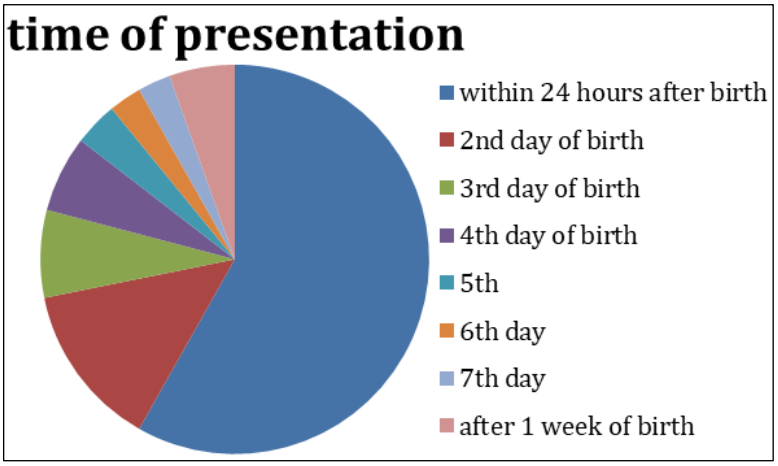

Fig 1: Time of Presentation

Table 1: level of ARM

Level	No of patients	No. of males	No. of females
High	50 (45.4%)	40	10
Intermediate	25 (22.7%)	20	5
Low	35 (31.8%)	10	25
Total	**110 (100%)**	**70 (63.6%)**	**40 (36%)**

Most of the patients had high level of defects (45.4%), followed by low level defects (31.8%). 27% had intermediate level of defects. High and intermediate type of defects were more common in males while low level of defects were more common in females.

Table 2: Associated anomalies

System involved	No. of patients
Urological	45
Sacro-spinal	24
Cardiac	60
Genital	30
Gastro-intestinal system	10
Limbs	8

Cardiac anomalies were the most commonly associated ones, followed by urological anomalies and genital anomalies. Most of the patients had anomalies of other systems as well.

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Table 3: Distribution of urological anomalies detected on ultrasound examination

Urological anomalies	No. of patients
Renalagenesis	25
Cystic disease of kidneys	6
Hydronephrosis	12
Duplexsystem	4
Ectopic kidneys	8
Total	45

Most of the patients with urological abnormalities had agenesis of kidneys, followed by hydronephrosis.

Table 4: Distribution of urological abnormalities as detected in VCUG

Abnormalities	No. of patients
Vesico-ureteric reflux grade - 1	1
Vesico-ureteric reflux grade - 2	0
Vesico-ureteric reflux grade - 3	1
Vesico-ureteric reflux grade - 4	1
Vesico-ureteric reflux grade - 5	37
Posterior urethral valves	5

All patients with abnormalities of urogenital system had undergone ultrasound examination of abdomen and VCUG was done in suspicious cases. Vesico-ureteric reflux was the most common urological finding on VCUG.

Discussion

Anorectal malformations (ARM) are often diagnosed at birth by the presence of imperforate anus. In this prospective study, 100 neonates with anorectal malformations were included and their clinical presentations and associated anomalies were analyzed.

In present study, males were most common than females. Many studies have shown a male preponderance in presentations of ano-rectal anomalies [10, 11].

In present study, high ARM are more common than low ARM's. High and intermediate level of ARM's is more commonly seen in males, while female have low level of ARM's.

The incidence of urogenital anomalies in present study is 68% which is similar to the study conducted by Sohail *et al*., [11] (62.2%), Mittal *et al*., [12] and Ahmed *et al*., [13].

Most common urological abnormality detected on ultrasonography was agenesis of kidney, followed by hydronephrosis. This is similar to study done by Stoll *et al*., [14]. The most common urological abnormality detected on VCUG was vesico-ureteric reflux. Similar observation of highest incidence of VUR was seen in studies done by Misra *et al*., [16]. According to Boemers [17], the incidence of VUR in patients with ARM is 19-47.2%.

Conclusion

Early identification and management of ano-rectal malformations is crucial as most of the patients have other associated anomalies which can be life-threatening. Routine cardiac and abdominal screening of neonates with ARM is necessary. A multidisciplinary approach consisting of pediatricians, pediatric surgeons, cardiologist, psychiatrists and radiologists is required to manage patients with anorectal malformations.

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Conflicts of Interest: No conflicts of interest declared.

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