Late Presentation of Anorectal Malformations - A Dilemma in Modern World: Cross-sectional Analysis from a Tertiary Care Hospital, Pakistan

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ABSTRACT

Objective: To study the presentation age of children with anorectal malformation and its relationship with complications.
Study Design: Cross-sectional study.
Place and Duration of Study: Children Hospital and Institute of Child Health, Faisalabad, from Jan to Dec 2019.
Methodology: A total of 170 patients were included in the study. All the infants with a positive diagnosis of anorectal malformation from birth to 2 years of age were included in the study. Variables, including age at presentation, gender, diagnosis, length of stay and outcome in the form of discharge or mortality, were recorded.
Results: A mean age of presentation was 3.4 ± 2.1 days. Out of the total 170 patients, 110 (64.71%) were males, and 60 (35.29%) were females. The age of children extended from birth to 2 years of age. It was observed that only 10 (5.88%) infants presented within 48 hours of birth, 110 (64.71%) neonates presented between 48 to 72 hours, while 50 (29.41%) presented after 72 hours. A mortality rate of 3.53% was observed in the late-presentation group, i.e., after 72 hours of birth. The length of hospitalization was longer in groups with late presentation.
Conclusion: The present study reported that the majority of the infants with anorectal malformation were admitted to the hospital with a late presentation, i.e., after 72 hours of birth. Early and timely presentation of these infants can improve patient outcomes and reduce mortality among infants with anorectal malformations.

Keywords: Anorectal malformation, Late presentation, Mortality.

INTRODUCTION

Medical Practitioners created an opening in children's perineum, those who suffered imperforate anus for several centuries. Unfortunately, the defect we recognize as "high" did not survive the treatment, while the children with "low" defects mostly did. Anorectal Malformations are one of the most common surgical problems that are faced by paediatric surgeons all over the globe.[1] They belong to a spectrum of diseases that can range from simple imperforate anus (low lying) to complete caudal regression.[2] Although antenatal ultrasound facilities are established to such an extent that these can be identified even in the prenatal period, several patients present late and with severe complications in our region.[3] Delayed presentation of these anorectal malformations in children leads to increased incidence of complications in the form of perforation, leading to septicemia that leads to high morbidity and mortality in return.[4-5]

The current study aimed to determine the incidence of delayed presentation of children with anorectal malformations and highlight the importance of early diagnosis concerning neonatal mortality and poor patient outcome.

METHODOLOGY

A cross-sectional study was conducted at the Children Hospital and Institute of Child Health, Faisalabad, from January 2019 to April 2020. The Children Hospital and Institute of Child Health is a tertiary care teaching level hospital that provides services to the entire district of Faisalabad. The daily out-patient department patient load is around 2000 patients per day, providing services to around 10 million population.

After taking ethical clearance from the Institutional Review Board of the Children Hospital and Institute of Child Health (reference number # 14/ 2020), the data collection was started. The non-probability consecutive sampling technique was used to enrol patients in the study. ARM prevalence was found to be 3.09% per 10000 births from the 4 litrarure.[6] By keeping a prevalence of 3.09%, a confidence level of 95%, and a margin of error of 3.08%, the sample size

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was calculated using "Select Statistics". A sample size of 170 was obtained.[6]

Inclusion Criteria: All the children who presented to the hospital with the complaint of not passing stool at birth, either through the Outdoor Patient Department or through the Emergency Department with the diagnosis of anorectal malformations, were included in this study.

Exclusion Criteria: Patients older than two years of age and female patients with rectovestibular fistula passing stool through fistulous opening were excluded from the study.

All the patients were clinically evaluated and X-ray invertogram was performed for each case. Those patients with high lying fistula underwent a diversion sigmoid colostomy, and those with low lying fistula underwent primary anoplasty. All the variables, including age at presentation, gender, diagnosis, an indication of surgery, preoperative and postoperative status, length of stay and outcome in the form of discharge, morbidity or mortality, were recorded in a predefined proforma. All the patients were divided into three subgroups according to the age at presentation, i.e. i) within the first 48 hours, ii) from 48 hours to 72 hours, and iii) more than 72 hours.

Statistical Package for Social Sciences (SPSS) version 24.0 was used for the data analysis. Categorical variables were presented as frequency and percentages. In contrast, the continuous variables, including the age at presentation and the length of hospitalization, were presented as mean ± SD.

RESULTS

The total number of patients included in the study was 170. Out of the total, 110 (64.71%) were males, and 60 (35.29%) were females. The age of children extended from birth to 2 years of age, with a mean age of presentation of 3.4 ± 2.1 days.

It was observed that only 10 (5.88%) infants presented within 48 hours of birth, 110 (64.71%) neonates presented between 48 to 72 hours, while 50 (29.41%) presented after 72 hours (Table).
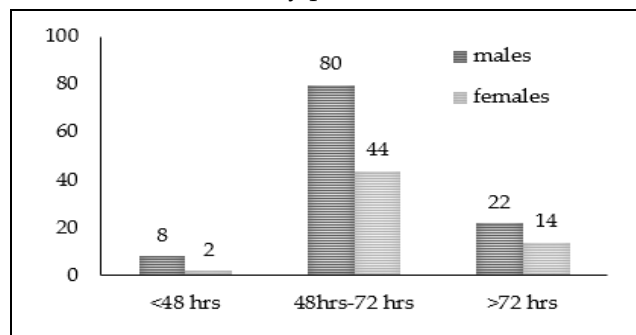
The gender-wise distribution according to the age at presentation was illustrated in Figure-1. It was observed that 8 (4.71%) males and 2 (1.18%) females presented within 48 hours of birth, while the majority of the male (80, 47.06%) and females (22, 12.94%) presented between 48 and 72 hours of birth. A total of 44 (25.88%) males and 14 (8.24%) females presented after 72 hours of birth.

**Table: Demographic characteristics of infants in the study (n=170).**

Characteristics	n (%)
Age at presentation	
Within 48 hours of birth	10 (5.88%)
Between 48 hours and 72 hours of birth	110 (64.71%)
After 72 hours of birth	50 (29.41%)
Gender	
Male	110 (64.71%)
Female	60 (35.29%)
Body Mass Index (BMI in kg/m2)	
Underweight	54 (31.7%)
Normal weight	116 (68.2%)
Overweight	-
Obese	-
Mortality	
Within 48 hours of birth	-
Between 48 hours and 72 hours of birth	-
After 72 hours of birth	6 (3.53%)

A total of 6 (3.53%) patients died during the study, and all infants who did not survive were presented after 72 hours of birth, i.e. late presentation. The length of stay and morbidity was also significantly low in those who presented within 72 hours compared to those who presented after 72 hours.

As illustrated in Figure-2, the mean length of hospitalization for infants with the late presentation, i.e. after 72 hours of age, was comparatively longer than those with the early presentation.

Figure-1: Gender-wise distribution according to the age at presentation (n=170).
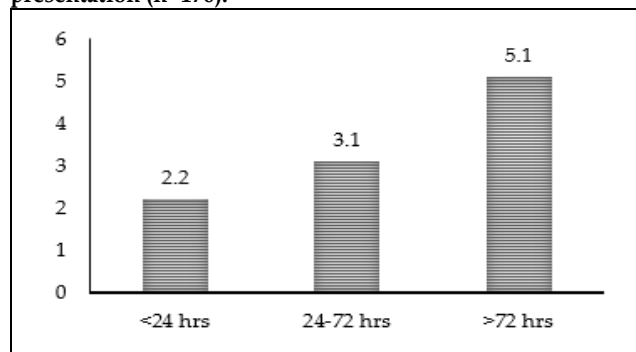
Figure-2: The mean length of hospitalization according to the age at presentation.

DISCUSSION

We reported a significant delay in presentation among infants with an anorectal malformation, which led to increased mortality. This needs to be addressed by educating the masses, including general practitioners and paediatricians so that the overall survival can be improved further.

One of the congenital abnormalities dealt with is the occurrence of anorectal malformations. However, if identified at an early stage, these are easily manageable and have a good patient outcome. The majority of these abnormalities are diagnosed over the clinical examination of the neonate. It is recommended to perform colostomy for babies at birth with high-type anorectal anomalies irrespective of gender.[2,6] The male babies presented with a low-type abnormality are benefitted from single-stage anoplasty performed at birth.[3] However, for female patients diagnosed with a low-type congenital abnormality, the majority of the surgeons prefer to treat the anomaly at presentation, which can be between birth and six months of age.

Fernández-Viadero *et al,* have reported that the majority of the babies were operated on between two to three months of age. Others concluded that it is beneficial to perform corrective surgeries soon after delivery of the baby to optimize the patient treatment and because the rate of complications in patients is lower.[7] More importantly, with time, much efficacious and safer methods for anaesthesia have been introduced, which lead to a better postoperative outcome. Another factor which leads to the considerable difference in patient outcomes is the optimized monitoring during surgery.

It has been established that earlier diagnoses of the anorectal birth abnormalities hold a significantly better prognosis than delayed diagnosis. The delayed diagnosis can adversely affect the treatment and management of these patients. There have been instances of delayed diagnosis or presentation; however, these instances are much higher in Pakistan, where the patient is presented much later in the disease course.[8,9]

In a study by Minaev *et al,* the outcome of 104 ARM cases was evaluated, which were managed with laparoscopy.[8] The mean age at presentation was 11.3 ± 0.4 months. In more than half of the patients, 39 (54.2%), good functional results were obtained. Perineum prolapse and tactile weakened anal reflex were found in 15.3% and 6.9% of patients.[8] None of the patients experienced poor outcomes as detected upon barium enema.

It is important to realize that an imperforate anus is diagnosed at birth or soon after birth with a physical examination.[9] Therefore, it is recommended that the neonate is examined at least twice or thrice from birth to discharge, initially at the delivery room upon birth, secondly at the nursery within 12 hours of birth, and at the time of discharge from the hospital.

In some cases, the diagnosis of ARM is made accidentally upon clinical examination. For instance, Kamat and Hrabovsky reported a case where a seven-month-old child could not pass a swallowed coin due to an imperforate anus that remained undiagnosed till then. This indicates the significance of adequate clinical examination of the urogenital area of a neonate upon birth to detect any physical abnormalities. Early diagnosis and management are the keys to a fair and good outcome in these children.[10]

In short, the standard protocol states that a newborn should be routinely examined within the first three days of life for any form of physical anorectal abnormality.[11,12] Furthermore, it is also advised that the perineum should be thoroughly observed and examined. Any suspicious abnormality or finding should be further investigated. The patency of the anus must be checked in the neonate at birth, even if meconium has been passed.[13,14]

Perforations in patients with anorectal malformation are rare and often occur after a delayed presentation.[15,16] The aetiology and the pathogenesis of perforation in neonates with ARM are explained by factors.[17] The downstream occlusion results in proximal intestinal dilatation and increased intraluminal pressure, resulting in tension gangrene. In various factors such as delayed presentation and absence of fistula, the gangrene progresses to a perforation, with the large bowel being the most common site.[18]

It has been shown that delayed presentation of children with anorectal malformation leads to increased incidence of morbidity in the form of increased length of stay and increased risk of perforation and leads to increased incidence of mortality.[4,5,19] Therefore, it is essential to educate the local masses in our society and sensitize the General Practitioners for early identification and referral of such children to avoid significant morbidity.

LIMITATIONS OF STUDY

Despite the many strengthening points of the current study, we still faced many problems during the study period. Due to the small sample size, statistical testing could not be performed. We hope that the current research can act as a

catalyst for future research and recommend that further studies should focus on acquiring a larger cohort of patients with a more thorough and diligent analysis.

CONCLUSION

The present study reported that the majority of the infants with anorectal malformation were admitted to the hospital with a late presentation, i.e. after 72 hours of birth. Early and timely presentation of these infants can improve patient outcomes and reduce mortality among infants with anorectal malformations.

Conflict of Interest: None.

Authors' Contribution

HS: Conception and design, TN: Critical review, and editing, MAF: Data acquisition and editing, JA: Data analysis and interpretation, MAN: Data acquisition, drafting, AA:, MA: Drafting of Article.

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