Section: Anatomy

Case Report

AN OVERBURDENED HEALTHCARE SYSTEM LEADING TO DELAY IN ANORECTAL MALFORMATIONS' DIAGNOSIS? A CASE STUDY AND COMPREHENSIVE REVIEW

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Abstract

One of the leading reasons for morbidity and mortality in developing nations is the failure to take neonates with anorectal malformations (ARM) to the hospital. Due to workload, a female infant whose mother had been complaining of constipation to the hospital regularly for the past 10 months received laxative drugs without a digital rectal exam. Around this time, her parents reported that she had been crying excessively and that it had been painful defecation. A small anus hole was revealed right near the vaginal fourchette on a rectal examination. Our case study figures out whether the source healthcare facility affects the timing of diagnosis as well as identifies modifiable elements that may quicken diagnosis and referral. The probability of missed anorectal malformation diagnoses needs to be considered in all individuals with severe and persistent constipation.

INTRODUCTION

Typical congenital defects of the terminal hindgut are anorectal malformations (ARM). The perineum should be carefully examined in order to identify ARM at birth or soon after. One of the main factors contributing to mortality and morbidity in developing nations is the delay in presenting infants with ARMs to the hospital. Nonetheless, more cases of newborns with ARM and a fistula have been documented, with additional delays in the proper diagnosis and treatment persisting well into adulthood.[1,2] There are 4 fundamental steps: Inspection, examining the anocutaneous reflex, digital rectal examination (DRE), and performing maneuvers to evaluate the anorectal condition. The majority of doctors reported lacking confidence in performing DRE or making diagnoses, but the majority of students believed their training was insufficient. Among reasons for not conducting DRE, it was claimed that the "patient's modesty," "extremely invasive," "limited utility," "convenient," and "gender/chaperone" was relevant. As a result, it is still difficult to properly train people to use DRE, which highlights the importance of education and training at all levels. Mannequin training greatly increased comfort level when doing DRE.[3] The essential need is for adequate education, a well-developed healthcare system, and training for health professionals in the early detection of ARM through a careful perineal examination of all neonates at delivery. Early lethal consequences like sepsis, intestinal perforation, and death are linked to diagnosing delays. Constipation and a large pubic region are late issues. In addition to changing surgical management, these problems can significantly worsen social and psychological morbidity. Even in multi-disciplinary teaching centers for primary care, the physical examination of newborns is still the initial step in detecting a disorder.[4,5]

We discuss the effects of failing to identify patients with ARM. Our case study intends to uncover modifiable factors that may hasten diagnosis and referral as well as to ascertain whether the source healthcare facility has an impact on the timing of diagnosis. The high rate of ARM diagnosis delays emphasizes the value of a thorough clinical evaluation of a newborn's perineum. The probability of missed anorectal malformation diagnoses needs to be considered in all individuals with severe and persistent constipation.

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CASE REPORT

A female infant was regularly taken to the hospital by her mother for the past 10 months with a complaint of constipation and consistently received laxatives medications without a digital rectal exam due to workload. About this time, the parents came in claiming that she would cry excessively every time she passed stool and that it had been painful. There were no respiratory problems and no significant family or psychiatric history. Rectal examination revealed a small anus orifice near the vaginal fourchette, as seen in [Figure 1]. The fourchette-to-coccyx distance was measured to be 3.7 cm, and the anal opening's center was observed to be 1.1 cm from the vaginal fourchette. The posterior rectal shelf was observed during a digital rectal examination. Additional congenital anomalies were not discovered. The results of the blood test, as well as the chest and abdominal radiographs, were inconsequential. The mother claimed that the child had a normal vaginal birth and had never been admitted to a neonatal intensive care unit. Breastfeeding continued until the patient was 6 months old. Before that, the child had a lot of trouble passing semi-liquid stools. The infant's supplemental feeding was initiated at 6 months. At birth, the anal deformity was undetected.

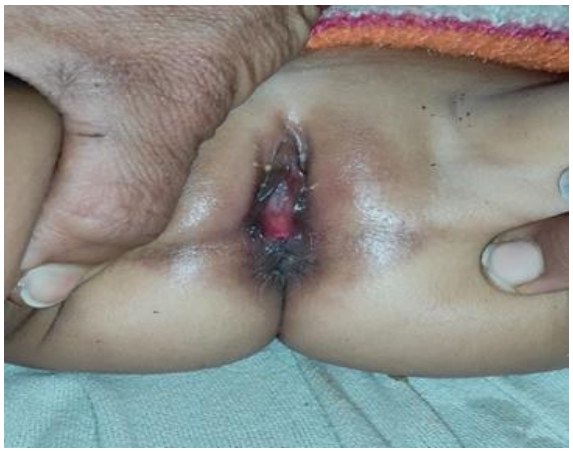

Figure 1: Perineal region shows a small anus orifice near the vaginal fourchette.

This case is diagnosed using the Anal Position Index. Reisner et al. (1984) provided a simple method for defining the normal position of the anus in newborns using the API (Anal Position Index), which is the ratio of the anus-fourchette distance to the coccyx-fourchette distance in females and the anus-scrotum distance to the coccyx-scrotum distance in males. The diagnosis of the anterior ectopic anus is made subjectively on inspection. They claimed that a male neonate's API of <0.46 and a female neonate's API of <0.34 indicated anterior displacement of the anus.[6]
Anal Position Index was determined in this case as:
Anus-Fourchette distance = 1.1 cm
Coccyx-Fourchette distance = 3.7 cm
Anal Position Index = Anus-Fourchette distance ÷ Coccyx-Fourchette distance
= 1.1/3.7 = 0.29
Here API of 0.29 indicates that the child has an Anterior Displacement of the Anus.

DISCUSSION

The anorectal region has an unusual developmental defect known as anterior displacement of the anus (ADA), which usually results in constipation but is not widely recognized. The actual prevalence of this disorder is not fully understood. The cloacal membrane is divided into the ventral urogenital and dorsal anal membranes by the embryologically posterior expansion of the perineal body. Ectopic anal openings can occur in the vestibule in females and the base of the scrotum in males due to failure or abnormalities in this division, as well as in the intrabulbar fossa of the spongy urethra. In a retrospective case review of 75 patients conducted by Lindley et al. (2006), it was discovered that children whose anorectal abnormalities had been detected later than usual experienced noticeably greater complications, including one death.[7] In order to highlight the time of anorectal malformations diagnosis and the mortality and morbidity associated with any delay, Haider et al. (2007) conducted research on 52 patients. 28 cases (or 53%) of delayed diagnosis of the abnormality were discovered.[2] Two fatalities (4%), both directly attributable to the missed diagnosis, were unfortunately associated with this delay and were associated with significant morbidity in 19% of the cases. Wilson et al. (2009) conducted a retrospective review. An Australian tertiary paediatric hospital referred 19 neonates with a delayed diagnosis of an ARM over the course of the study's 7.5-year duration, which allowed researchers to analyze the prevalence and presenting characteristics of these infants. 14 (32%) of the 44 patients who were referred to our facility had an ARM diagnosis that was delayed by more than 24 hours. Obstructive symptoms were reported by a considerably higher percentage of patients with a delayed diagnosis (86%), including abdominal pain (35%), delayed passage of meconium or stool (29%), and abdominal distension (57%). Despite receiving a newborn assessment, 12 patients did not have ARM correctly identified. It appears that the current recommendations are insufficient to guarantee a fast diagnosis of ARM.[8] In a descriptive cross-sectional study by Lawal et al. (2012) a total of 365 mothers participated in the study, which sought to determine how well-informed they were about ARM and whether or not the anus appeared normally or abnormally. An educational intervention aimed at young mothers from low socioeconomic classes is suggested in the study.[9] In order to ascertain whether the source healthcare facility has an impact on the timing of diagnosis and to discover modifiable factors that may hasten diagnosis and referral,

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Govender et al. (2016) conducted a retrospective analysis, it is known that patients with anorectal malformations (ARM) have a significant risk of morbidity and mortality if diagnosis or referral is delayed. Indicated that staff education is required, especially in CHCs, on standard neonatal assessment and typical perineal anatomy. Girls, who frequently have decompressive fistulas, are particularly at risk for delayed diagnosis. Increased morbidity and mortality result from delays.[4] Overview of Anorectal Malformations in Africa by Lawal (2019) focuses on the epidemiology, clinical presentation, therapy, and outcomes of care for infants in Africa who have anorectal malformations and offers solutions used by pediatric surgeons who are dealing with limited resources.[10] To evaluate the rate of delayed diagnosis and related intestinal perforation in ARM, Kruger et al. (2019) performed a retrospective study on 243 ARM patients. Mention that a risk factor for consequences, such as intestinal perforation, is a delayed diagnosis. They found that in two individuals with delayed diagnosis, there was intestinal perforation and delayed diagnosis in ARM patients continues to be prevalent and even lethal.[1] In order to assess the presenting features and consequences of delayed versus early presentation, Reddy et al. (2021) conducted a prospective observational study to determine the effect of delay in the diagnosis of anorectal malformation (ARM) in neonates. revealed that almost half of the infants with ARM presented later than expected. Due to decreased weight at presentation, a higher frequency of abdominal distension, and infection, the delayed group required more time for resuscitation and more inotropes prior to surgery. Early surgical outcomes, including stoma function, feed initiation, and complete feed time, were markedly slowed in late presenters.[5] A case of late-diagnosed small congenital anorectal malformation was presented by Soeselo et al. (2020). Normal physical examinations at birth frequently fail to detect congenital anorectal malformations, which are typically simple to diagnose. Consequently, the missed-diagnosed abnormality causes the patient to present with substantial problems.[11]

Inspection, grading the anocutaneous reflex, conducting maneuvers to assess the anorectal state, and digital rectal examination (DRE) are the four basic steps. Yet, the majority of students felt their training was insufficient. The majority of doctors reported lacking confidence in performing DRE or making diagnoses. Among reasons for not administering DRE, it was claimed that the "patient's modesty," "extremely intrusive," "limited value," "convenient," and "gender/chaperone" was relevant. Because of this, it is still challenging to teach individuals how to use DRE effectively, which emphasizes the significance of education and training at all levels. The comfort level when doing DRE was considerably raised by mannequin training.[3,12] An effective healthcare system, proper information, and training for medical professionals in the early detection of ARM through a meticulous perineal examination of all newborns at birth are key requirements. Many children are not recognized early or undergo strenuous colonic cleansing routines by their doctors before being referred to a surgeon for the therapy of constipation because of this lack of identification combined with overburdened hospitals.

CONCLUSION

Healthcare facilities urgently need to recruit additional staff members with a focus on developing super-specialty hospitals and recruiting staff by creating doctor-friendly environments. Standard clinical protocols also need to be reviewed and updated to ensure that congenital or other defects are not overlooked and that diagnosis time is optimized.

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