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Delayed Diagnosis of Congenital Anorectal Malformations: A Plea for Standardized Neonatal Anorectal Examination

Alshehri A and Altokhais T*

College of Medicine, King Saud University Medical City, King Saud University, Saudi Arabia

***Corresponding author:**

Tariq Altokhais,
Associate Professor, Consultant Pediatric Surgeon, Division of Pediatric Surgery, Department of Surgery, College of Medicine, King Saud University, Saudi Arabia, Tel: +966-11-8066421,
E-mail: taltokhais@ksu.edu.sa

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1. Abstract

1.1. Background: Anorectal Malformation (ARM) includes a spectrum of different anomalies. Delayed diagnosis of the ARM type may lead to significant morbidity. We aim to analyze delayed or mismanaged ARM cases referred to a single hospital and explore reasons of mismanagement.

1.2. Methods: A retrospective review was conducted of patients with delayed presentation of ARM from March 2009 to September 2018. Delayed presentation was defined as patients not diagnosed with ARM in the first 48 hours of life or diagnosed with the incorrect ARM type. Basic demographic data, initial and final diagnosis, and management data were collected.

1.3. Results: 13 patients were analyzed: 10 (76.9%) were male, four (30.8%) had minor associated anomalies. All patients had perineal fistula, however, incorrect diagnosis included five patients (38.5%) as normal anus, three (23.1%) as recto-urinary fistula, three (23.1%) as anal stenosis, and two (15.4%) as anterior anus. Patients' presentation to our hospital was due to severe constipation in seven patients (53.8%), suspicion of abnormal anus in two (15.4%), or intestinal obstruction in one (7.7%). All patients underwent Posterior Sagittal Anorectoplasty (PSARP). Reasons of mismanagement included incorrect diagnosis by a pediatric surgeon in six patients (46.2%), a pediatrician in three (30.8%), or family socio-economic reasons in three (23.1%).

1.4. Conclusion: Delayed recognition of ARM often leads to significant morbidity to the patients. Surprisingly, pediatric surgeons are responsible for incorrect identification for most of our patients followed by pediatricians. Therefore, more effort should be made to standardize neonatal anorectal examination for the training doctors in pediatrics and pediatric surgery.

2. Introduction

Anorectal Malformation (ARM) comprises a wide spectrum of congenital anomalies that affect the anorectum and occasionally the adjacent genito-urinary system. It is of paramount importance to correctly identify the type of ARM before treatment is initiated. Different classification schemes were developed including the historic Wingspread classification that divided ARM into low, intermediate, or high type. In 2005, the Krickenbeck classification was developed by an international group aiming to standardize the diagnosis and treatment of the different types of ARM [1]. Nowadays, it is the most practical and widely used classification system for ARM [2-4].

Detailed neonatal anorectal examination remains the most important tool to diagnose ARM shortly after birth. Nonetheless, delayed recognition of ARM or incorrect identification of the type of anomaly still represents a significant problem that often result in suffering of the child and his/her family and perhaps mortality [5, 6, 15–21, 7–14].

The aim of this study is to analyze delayed or mismanaged cases of ARM referred to a single tertiary hospital and explore reasons of mismanagement.

3. Materials and Methods

A retrospective review was conducted of all the patients with delayed presentation of ARM to a single referral hospital in Riyadh,

Saudi Arabia from March 2009 to September 2018. Delayed presentation was defined to include patients who were not diagnosed with ARM in the first 48 hours of life or were diagnosed with the incorrect type of ARM, therefore received incorrect surgical management. Malformations were classified according to Krickenbeck classification [1]. Data collected included age, sex, gestational age, location of delivery, associated anomalies, diagnosis of ARM at the first 48 hours, initial treatment, age and symptoms at presentation to our hospital, final diagnosis and treatment provided. We also examined the reasons behind mismanagement and divided that into: incorrect diagnosis by the pediatrician, incorrect diagnosis by a pediatric surgeon, or family socio-economic reasons.

Descriptive statistics were generated using IBM SPSS Statistics for Mac, Version 20.0 (IBM SPSS Statistics, IBM Corp., Armonk, NY, USA). No comparative statistical tests were performed due to small sample size.

4. Results

During the study period, 13 patients were identified and analyzed. (Table 1) shows the baseline characteristics of our cohort. All patients were born at term, 10 (76.9%) were male, and four (30.8%) had minor associated anomalies including Ventricular Septal Defect (VSD), Posterior Urethral Valve (PUV), congenital cataract, and ear malformation. No major associated anomalies were identified. Nine patients (69.2%) of the cohort were Saudi nationals, seven (53.8%) were delivered in governmental hospitals, five (38.5%) in private hospitals, while one patient was delivered outside the country.

Although all patients had ARM with perineal fisulae, five (38.5%) were incorrectly diagnosed as normal anus, two (15.4%) as anterior anus, and three (23.1%) as anal stenosis managed with anal dilations. Interestingly, two patients were told to have a normal anus despite that both of them had a clear bucket handle deformity. Three patients (23.1%) were incorrectly diagnosed as recto-urinary fistula and underwent colostomy creation.

Patients' presentation to our hospital was due to severe refractory constipation in seven (53.8%) patients, suspicion of abnormal anus in two (15.4%), or intestinal obstruction in one (7.7%), as detailed in (Table 2). Mean age at presentation was 6.4 months ranging from three days to 19 months. Once the correct diagnosis was made, all patients underwent Posterior Sagittal Anorectoplasty (PSARP) in our hospital without any major complications. Median follow up was four months.

(Table 3) shows reasons of mismanagement included incorrect diagnosis by a pediatric surgeon in six patients (46.2%), incorrect diagnosis by a pediatrician in four (30.8%), or family socio-economic reasons in three (23.1%). All relevant patient's details are summarized in (Table 4).

Table 1: Baseline characteristics of the patients

Baseline Characteristics	
Gestational age (mean in weeks)	39.3
Sex: (n,%)	
- Male	10 (76.9%)
- Female	3 (23.1%)
Nationality: (n,%)	
- Saudi	9 (69.2%)
- Non-Saudi	4 (30.8%)
Location of delivery: (n,%)	
- Governmental hospital	7 (53.8%)
- Private hospital	5 (38.5%)
- Outside the country	1 (7.7%)
Associated congenital anomalies: (n,%)	
Minor	4 (30.8%)
Major	0
Initial diagnosis after birth: (n,%)	
- Normal anus	5 (38.5%)
- Anal stenosis	3 (23.1%)
- Anterior anus	2 (15.4%)
- Imperforate anus with no perineal fistula (possible recto-urethral fistula)	3 (23.1%)
Management after birth: (n,%)	
- None	8 (61.5%)
- Anal dilations	2 (15.4%)
- Colostomy	3 (23.1%)

Table 2: Data of the patients at the time of presentation to our hospital

Age (mean in months)	6.4
Reason for presentation: (n,%)	
- Constipation	7 (53.8%)
- Referred for possible recto-urinary fistula	3 (23.1%)
- Suspicion of abnormal anus	2 (15.4%)
- Intestinal obstruction	1 (7.7%)
Correct diagnosis: (n,%)	
- Perineal fistula	13 (100%)

Table 3: Reasons for the delayed diagnosis

Reasons for the delayed diagnosis: (n,%)	
- Incorrect diagnosis by a pediatric surgeon	6 (46.2%)
- Incorrect diagnosis by a pediatrician	4 (30.8%)
- Family socio-economic status	3 (23.1%)

Table 4: Summary of the patients' data

Case No.	Sex	Initial Diagnosis	Initial Management	Cause of Delayed Presentation	Age at Presentation	Presenting Symptom (s)	Correct Diagnosis	Associated Anomalies	Surgical Management
1	Male	Normal anus	None	Missed diagnosis by a pediatrician	3 days	Vomiting, abdominal distension	Perineal fistula	None	PSARP
2	Male	Anterior anus	Anal dilations	Family socio-economic reasons	10 days	Suspicion of abnormal anus	Perineal fistula	VSD	PSARP
3	Male	Anal stenosis	None	Missed diagnosis by a pediatric surgeon	6 months	Constipation	Perineal fistula	None	PSARP
4	Female	Anterior anus	None	Family socio-economic reasons	25 days	Suspicion of abnormal anus	Perineal fistula	None	PSARP
5	Male	Normal anus	None	Missed diagnosis by a pediatric surgeon	19 months	Constipation	Perineal fistula	None	PSARP
6	Female	Recto-urethral fistula	Colostomy	Missed diagnosis by a pediatric surgeon	9 months	Referred for treatment of recto-urethral fistula	Perineal fistula	Congenital cataract	PSARP
7	Female	Anal stenosis	Anal dilations	Missed diagnosis by a pediatric surgeon	9 months	Constipation	Perineal fistula	None	PSARP
8	Male	Normal anus	None	Missed diagnosis by a pediatrician	5 months	Constipation	Perineal fistula	Ear malformation	PSARP
9	Male	Recto-urethral fistula	Colostomy	Missed diagnosis by a pediatric surgeon	3 months	Referred for treatment of recto-urethral fistula	Perineal fistula	PUV	PSARP
10	Male	Normal anus	None	Missed diagnosis by a pediatrician	3 months	Constipation	Perineal fistula	None	PSARP
11	Male	Recto-urethral fistula	Colostomy	Missed diagnosis by a pediatric surgeon	3 months	Referred for treatment of recto-urethral fistula	Perineal fistula	None	PSARP
12	Male	Anal stenosis	None	Family socio-economic reasons	14 months	Constipation	Perineal fistula	None	PSARP
13	Male	Normal anus	None	Missed diagnosis by a pediatrician	10 months	Constipation	Perineal fistula	None	PSARP

5. Discussion

The majority of anorectal malformations can be diagnosed readily after birth, however, some cases may be more challenging owing to unclear perineal morphology resulting in delay in diagnosis. It has been estimated that nearly half of the patients presenting to referral hospitals had delayed diagnosis of their ARM [12]. Significant morbidity and sometimes mortality are associated with the delay in management of such conditions [12–14, 22–24]. The definition of delayed cases remains controversial. Some authors have defined that as patients not diagnosed with ARM within the first 24 hours of life, but we think that may not be practical because it often requires at least 6-24 hours for the distal rectum to distend and meconium to appear in the perineum indicating a perineal fistula [6, 10, 11, 14, 18]. Other groups have defined delayed cases as those diagnosed after 48 hours of life or even after the newborn period [13, 16, 21]. In our study, we present a commonly underreported cohort of patients with prolonged but preventable suffering. We have shown that our patients had waited, on average, six months before having their definitive surgery. The commonest symptom was refractory constipation that had led most of these patients to be seen by several physicians. There was a relative male predominance in our cohort, which is different from previous reports that suggested female predominance among delayed cases likely due

to relatively more difficult examination [18, 21]. Once the diagnosis of perineal fistula was confirmed, all of our patients underwent PSARP without complications. Some surgeons may elect to perform colostomy for perineal fistula followed by staged repair particularly with significant associated anomalies, however, we did not think that was necessary in our patients as the functional outcomes are usually excellent after primary PSARP [21].

We were surprised to see that nearly half of our cohort was diagnosed to have a normal anus whereas they in fact had perineal fistula. Two of these patients had bucket handle deformity presented at the 3rd day and 19th months of age. What was alarming to us is that nearly half of our patients were misdiagnosed by pediatric surgeons in community practice.

The importance of careful neonatal examination cannot be overemphasized. A good examination should be performed with adequate lighting and after the meconium, if any, is cleaned. A normal anus should fulfill 2 main criteria: 1- it should be located within the center of the sphincter muscle complex, which is often covered, by a slightly darker skin. 2- it should be large enough to allow passage of a 10-12 mm Hegar dilator for a term newborn (this is merely based on expert opinion). 2-. If there is doubt in the diagnosis, the surgeon should have a low threshold to perform examination under anesthesia with muscle stimulation. The presence of bucket handle deformity, sometimes coupled with meconium pearls, is almost always associated with perineal fistula [3, 4, 25].

Our study was limited by its small sample size from a single tertiary hospital. However, we feel that this group of patients is often underreported both nationally and internationally. Another limitation is the unclear explanation of the reasons behind failing to establish a diagnosis after birth.

In this study we highlighted some important features of patients presenting with delayed diagnosis of ARM. This delay often results in prolonged suffering of the patients and their families. We should increase the awareness about the importance of a good quality anorectal examination particularly in neonates in general and older children with significant constipation. Additionally, particular emphasis should be placed on standardizing anorectal examination for the training doctors in pediatrics and pediatric surgery.

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