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Case Report

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A female neonate with absent anal opening and dug perineum: Think high

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ABSTRACT

Background: A female neonate with anorectal malformation (ARM) may have one, two, or three openings in the perineum. One opening represents cloaca while three openings usually suggest low ARM. Females with two openings in the perineum may be ARM without any fistula, rectovaginal fistula, or absent vagina. The association of ARM with multiple intestinal atresias and malrotation is rare.

Case Presentation: We present here a case of a female neonate with two perineal openings, whose perineum was explored by a general surgeon without any radiological investigation and was later found to have a high ARM, multiple jejunal and ileal atresia with malrotation.

Conclusion: ARM associated with small bowel atresia and malrotation are rarely described. Such cases require management in Pediatric surgery settings.

INTRODUCTION

The incidence of anorectal malformations (ARM) is approximately 1 in 5000 live births, while that of jejunoileal atresia ranges from 1 in 5000 to 14,000 live births.[1,2] Approximately 50% of neonates with ARM have one or more other abnormalities. VACTERL (vertebral, anorectal, cardiac, tracheal, esophageal, renal, and limb) association needs to be ruled out. ARM associated with jejunoileal atresia has rarely been described in the literature.[3,4] Rarer still is its coexistence with malrotation. Herein we describe a case of ARM associated with jejunoileal atresia and malrotation of the intestine.

CASE REPORT

A full-term female neonate (birth weight 2.1kg), born by spontaneous vaginal delivery at a primary community health center, was attended by a general surgeon for the imperforate anus, who then explored the perineum (without any radiological investigations) but could not identify rectal pouch thus closed the perineal wound and referred the baby to Pediatric surgeon.

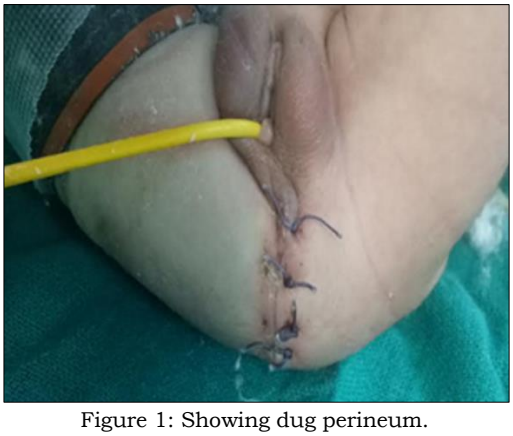

Figure 1: Showing dug perineum.

On admission to our center on day 5 of life, the general condition was sick. The anterior fontanelle was sunken. Capillary refilling time was >3 seconds with loss of skin turgor. Her heart rate was 144/min, systolic blood pressure was 54 mm Hg, and a respiratory rate of 35/min. Her upper abdomen was distended. The perineum had two openings (urethra and vagina). The anal opening was absent with a recently sutured midline perineal wound (Fig.1). No other gross malformations were apparent on general physical examination.

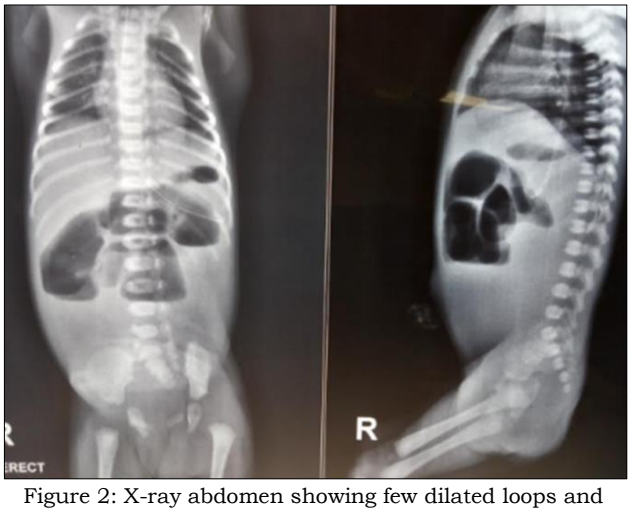

Figure 2: X-ray abdomen showing few dilated loops and air-fluid levels.

The baby was initially stabilized with intravenous fluid boluses and triple antibiotics were started. The baby was catheterized and a nasogastric tube was passed and 20 ml bilious fluid aspirated. After stabilization, an abdominal radiograph was done that demonstrated a few dilated small bowel loops in the central abdomen with air-fluid levels (Fig.2). Routine blood investigations were normal except for raised urea (48 mg/dl) and creatinine (0.8 mg/dl). The baby was planned for laparotomy and revealed a type IV intestinal atresia as well as malrotation (DJ to right of the spine). Three atresias were found, the first 25 cm distal to DJ flexure, second 14 cm distal to the first atresia, and third 15 cm proximal of ileocecal junction (Fig.3).

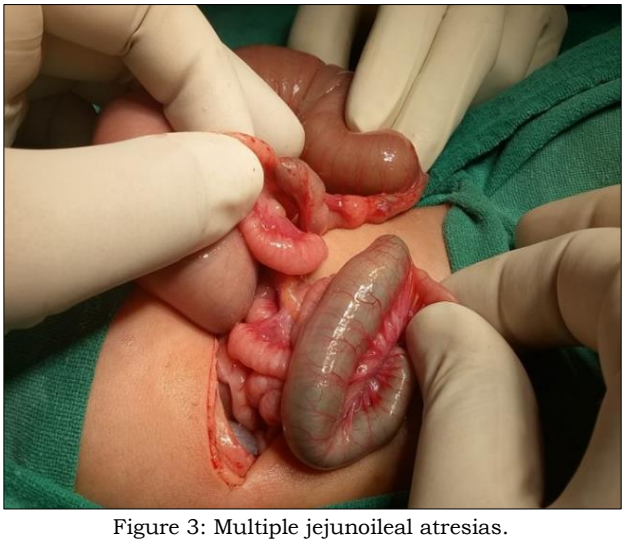

Figure 3: Multiple jejunoileal atresias.

For malrotation, the duodenum was kocherized and mesentery was broadened. Resection and anastomoses were done for small gut atresias. The proximal anastomosis was done after resection of proximal two atresias and second at distal ileum approximately 10 cm proximal to the ileocecal junction with diversion high sigmoid colostomy for high ARM (HARM). The remaining length of the small bowel was about 65 cm with an ileocecal valve. Postoperatively, the patient recovered well in the neonatal intensive care unit and received total parenteral nutrition. Blood culture sent at admission had Candida tropicalis growth and received 2 weeks of fluconazole as per sensitivity. Bowel movement started after about the 3rd postoperative day and after 2 weeks, she tolerated complete nutritional support orally. The ultrasound did later show bilateral normal kidneys and normal 2D echocardiography. The baby is awaiting definitive surgery for HARM.

DISCUSSION

Malrotation is an anatomical entity of intestinal malposition affecting 0.2 to 1% of the general population.[2] It is a congenital anomaly that may cause midgut volvulus on a narrow-based mesentery and may lead to antenatal type III atresia. It is present in about 20% of jejunoileal atresias.[2] The intestinal malrotation may be an integral part of anterior abdominal wall defects or diaphragmatic hernias, but its concomitant association with ARM and intestinal atresia is rare.[5] Powell et al studied 70 patients of malrotation over 15 years, out of whom 6 had imperforate anus and 5 jejunal atresias.[6]

A few cases of ARM associated with jejunoileal atresia have been reported in the literature.[3,7,8,9] Concurrent intestinal atresia in patients of ARM is thought to result from vascular disturbances during embryogenesis. Intestinal atresias are often suspected in the antenatal period, as characterized by bowel dilatation and polyhydramnios in the 3rd trimester of pregnancy. However, this is not highly sensitive or specific because other conditions, including the imperforate anus, can present with similar prenatal ultrasound features. In our case, no antenatal ultrasound record was available. Postnatally, the baby had absent anal opening and progressive upper abdominal distension, and bilious aspirates pointing toward associated proximal bowel obstruction / atresia. In our patient, ARM was initially addressed by a general surgeon but due to the high variety of ARM, the rectal pouch might have not been identified via a perineal route.

A tabular representation of various available published reports of ARM with small intestinal atresias has been shown in Table 1. As noted, the outcome is generally good if complex cardiac anomalies or VACTERL association is ruled out. Mitra et al reported short bowel syndrome (SBS) in an operated case of HARM with multiple jejunoileal and colonic atresia and the length of the remaining bowel was 25 cm.[8] In our case, about 65 cm of the small intestine was available with preserved ileocecal valve, thus minimizing the risk of SBS. The need for nutritional support and total parenteral nutrition

cannot be underestimated in case of surgical SBS or failure to thrive. The index case is gaining weight in the follow-up period (2 months of age) and had no episodes of diarrhea.

Sinha et al gave the term "dug perineum" to describe ARM patients with unsuccessful attempts at perineal route repair. This usually happens due to attempts by general surgeons or a low index of suspicion at presentation. Second perineal surgery is difficult and has a poor outcome. It is associated with a high chance of urethral injury in view of dense fibrosis and loss of tissue planes. Post-operative functional outcome is also poor as compared to a primary posterior sagittal route surgery.[10] It is imperative to provide basic Pediatric Surgery training during residency/training for general surgery, so that proper management may be done, dug perineum avoided or at least expertise of Pediatric surgeon may be sought for various surgical congenital malformations.

In conclusion, this case represents a rare co-existence of HARM with multiple jejunoileal atresias and malrotation. The coexistence of these conditions can obscure the evaluation and illustrate the need for a high index of suspicion. This report also highlights the importance of inculcating Pediatric surgery training in the residency program of general surgery

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Table 1: Showing Published cases of anorectal malformations associated with small bowel atresia/stenosis.

Authors	Age/Sex	ARM type	Findings	Surgery	outcome
Asabe et al, 1997	1d/M	LARM	Ileal atresia	Septectomy + transverse colostomy	Survived
Asabe et al, 2004	1d/F	HARM	Sigmoid atresia + ileal stenosis	Stenosis correction + sigmoid colostomy	Survived
Maitra et al, 2012	2d/M	HARM	Jejuno-ileal atresia + colonic atresia	EEA + colostomy at colonic atresia site	SBS
Puri et al, 2016	1d/M	LARM	Ileal atresia	Resection of atretic bowel + double-barrel ileo-colostomy	Survived
Sunil et al, 2019	3d/M	Perineal fistula	Jejunal atresia	EEA + loop sigmoid stoma	LAMA

SBS: Short bowel syndrome, LARM: Low anorectal malformations, HARM: High anorectal malformations, LAMA: Left against medical advice, EEA: End to end anastomosis

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