2021

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International Journal of Surgery Case Reports 84 (2021) 106159

Contents lists available at ScienceDirect

International Journal of Surgery Case Reports

journal homepage: www.elsevier.com/locate/ijscr

Case report

Anorectal malformation, associated with colon atresia and intestinal malrotation, a case report

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ARTICLE INFO

Keywords:
Anorectal malformation
Colonic atresia
Malrotation
Diagnosis
Case report

ABSTRACT

Introduction and importance: In this case report from Muhimbili National Hospital, Dar es salaam, Tanzania, we present the unexpected findings of anorectal malformation, colonic atresia, and intestinal malrotation in a 2-day old neonate. This combination is exceedingly rare, with only case reports published in the literature. We describe the challenges in diagnosis and offer our insights based on this experience and review of the literature.

Case presentation: Our patient was a male born at term, weighing 2600 g, diagnosed clinically with a high anorectal malformation. He was planned for colostomy, and we unexpectedly found a collapsed descending colon. Exploration revealed intestinal malrotation and three segments of type I colonic atresia from the mid transverse colon to the sigmoid colon in addition to the high anorectal malformation.

Clinical discussion: Creating a colostomy in a high anorectal malformation and failure to identify proximal intestinal atresia would result in potentially devastating consequences. Colonic atresia and anorectal malformation will both present as large bowel obstruction. In the extremely rare situation, when occurring in combination, the obvious clinical diagnosis of anorectal malformation will mask the clinical suspicion of the possibility of colonic atresia. Finding a distal bowel air bubble above the pubococcygeal line on an invertogram is useful in identifying proximal atresia preoperatively.

Conclusion: The current report emphasizes the importance of maintaining an awareness of possible associated colonic atresia in neonates with anorectal malformation. An invertogram and intraoperative finding of a collapsed descending colon should prompt evaluation for a proximal obstructing lesion.

1. Introduction

Anorectal malformations (ARM) are among the more common major congenital anomalies accounting for around 1 in every 5000 live births [1] due to defects in the embryological development of the hindgut from the cloaca. The diagnosis can be made by demonstrating features of large bowel obstruction, failure of passage of meconium and imperforate anus [2]. Associated anomalies involving other organ systems constituting the VACTREL association exist.

The incidence of intestinal malrotation is 1 in 6000 live births, occurs in a spectrum due to a combination of incomplete intestinal rotation, retraction, and fixation, predisposing the infant to midgut volvulus. This condition is asymptomatic; however, an otherwise healthy child may present suspicious signs of midgut volvulus [3]. Plain abdominal X-rays, upper gastrointestinal contrast studies, and ultrasound establish the diagnosis [3,4]. Intestinal malrotation is associated with several other intestinal and extra-intestinal pathologies [3].

On the other hand, colon atresia (CA), which occurs due to in utero mesenteric vascular insults, is an extremely rare congenital malformation accounting for 1 in every 20,000 live births [5,6]. Like ARM, CA presents as large bowel obstruction with failure to pass meconium, but in contrast to ARM, with a patent anus. Plain X rays will demonstrate large bowel obstruction, and contrast enema may demonstrate the level of the atresia [7]. This is associated with other anomalies in about 30% of the cases [7–9].

The findings of a high ARM, CA and intestinal malrotation, three embryologically separate non-associated congenital anomalies of the gastrointestinal tract in the same neonate, are rare with only case reports published and pose significant diagnostic challenges [10]. In this case report from Muhimbili National Hospital, Dar-es-Salaam, Tanzania, we

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https://doi.org/10.1016/j.ijscr.2021.106159
Received 6 June 2021; Received in revised form 25 June 2021; Accepted 26 June 2021
Available online 30 June 2021

describe the unexpected intraoperative findings, the challenges in diagnosis, and our insights based on this experience and review of the literature. This paper has been reported in line with the SCARE 2020 criteria [11]. This article has been registered with the Research Registry with identification number researchregistry6801 and can be found through the following hyperlink Browse the Registry - Research Registry and has ethical clearance from Muhimbili National Hospital institution review board with reference number MNH/IRB-CR/2021/001.

2. Case presentation

2.1. Introduction and background

A 2-day-old male referred to us from a peripheral health centre, born at term via spontaneous vaginal delivery weighing 2600 g and reported to have cried immediately with an APGAR score of 8 and 10 at 1 and 5 min, respectively. The pregnancy was uneventful without febrile illnesses or exposure to known teratogenic agents. Antenatal scans were not done. At birth, the baby was found to have an imperforate anus without meconium in urine or in the perineum. Initial resuscitation and nasogastric tube (NGT) decompression was placed before being referred to our hospital at 48 h since birth.

2.2. Clinical and diagnostic assessment

He was alert, not pale, or jaundiced, with a pulse rate of 131, respiratory rate of 44, maintaining saturation in room air, with a tense generalized abdominal distension. The perineal examination was significant for the imperforate anus, without perineal fistula with well-formed gluteal cleft and anal dimple (Image 1 – perineal examination findings Fig. 1). The rest of the newborn examination was normal. The clinical impression was of large bowel obstruction due to ARM, most likely an intermediate or high type. Work up revealed Hb- 18.1 g/dL, PLT- 251 k/uL, WBC- 4.4 k/uL, Na- 137 mmol/l K- 3.5 mmol/L and Cl- 98 mmol/L. Echocardiography revealed a structurally normal heart. Kidney, ureter, bladder and spine USS were not done.

Given that 48 h had elapsed since birth and features suggestive of high ARM, a staged management approach of an emergency de-functioning colostomy first was deemed appropriate.

2.3. Therapeutic information

Following counseling and informed consent from the mother, a senior pediatric surgeon carried out the procedure under general anaesthesia. The initial incision was transverse left iliac fossa. Unexpectedly, the descending colon was fixed to the retroperitoneum, not dilated, and could not be delivered through the incision for descending double-barrel colostomy. On the other hand, the cecum was grossly dilated and mobile, raising a suspicion of an obstructing lesion proximal to the descending colon.

Laparotomy via a transverse supraumbilical incision was made to explore the peritoneal cavity fully. Findings revealed mal-rotated small bowels characterized by an absence of ligament of Treitz, a duodenal jejunal junction on the right side, with narrow-based small bowel mesentery, Ladd's bands, without associated volvulus. The cecum was mobile, distended to the level of the mid transverse colon with a sharp transition zone, collapsed colon distal to this level, with two other occlusions at the splenic flexure and descending colon. There were no associated gaps between the intestinal segments or mesenteric defects on the transition zone, fitting the descriptions of Type I Bland-Sutton-Louw classification of colonic atresia (Image 2- Laparotomy findings Fig. 2). The malrotation did not contribute to the clinical presentation but was managed due to a persistent risk of developing midgut volvulus post-operatively.

Lysis of Ladd's bands was done to widen the small bowel mesentery, resulting in the location of the cecum on the left iliac fossae, additionally appendectomy was also performed. Attempts at definitive management of the atretic segments were deferred due to the emergency nature and the intent of conducting a short, abbreviated damage control surgery. A de-functioning double-barrel colostomy was created proximal to the atretic colonic segments and exteriorized at the initial left iliac fossae incision.

Post-operative, he was kept on IV ceftriaxone 100 mg daily, IV paracetamol 30 mg 8 hourly and IV neonatal cocktail fluid to meet both maintenance and suspected losses. The baby was transferred to the NICU for close monitoring. Seventy-two hours post-operatively, the baby succumbed. The exact cause was not determined as physiological derangements were not detected timely.

3. Discussion

Creation of a colostomy in a high ARM and failure of identifying proximal intestinal atresia would result in persistent intestinal obstruction post-operatively [12]. This situation has been associated with a mortality rate of more than 60% on the delay of diagnosis beyond three days. This occurs due to a closed-loop obstruction from a competent ileocecal valve and the atretic portion, rapidly progressing to intestinal perforation and intra-abdominal sepsis [7].

Small bowel intestinal atresia with ARM may be suspected in utero by ultrasound in the presence of polyhydramnios and clinical- radiologically after birth. Hence in this rare instance, a carefully evaluated neonate with small intestinal atresia and a concurrent ARM, the two diagnoses may be suspected preoperatively [13,14]. Early bilious emesis, upper abdominal distension, and abdominal x-ray showing features of upper gastrointestinal obstruction are features suspicious of small bowel atresia in the presence of ARM [13]. On the contrary, in CA and concurrent high ARM, the clinical-radiological evaluations are less discriminatory. The obvious clinical diagnosis of ARM will mask the clinical suspicion of a CA. This situation has resulted in the creation of unnecessary colostomies, relaparotomy and mortality [15].

Following the diagnosis of a high ARM, practice dictates the placement of an emergency de-functioning colostomy as the initial procedure [2]. During the pre-operative evaluation, the finding of the distal air bubble above the pelvis in an invertogram should raise the suspicion of colonic atresia [15,16]. In our case, an invertogram was omitted due to the unawareness of the existence of concurrent ARM and CA. A case report from Japan of an ARM with ileal atresia cited the importance of examining the entire alimentary tract when an invertogram reveals bowel gas above the pubococcygeal line [15]. A similar recommendation was made from a case in Greece [16]. The cross-table lateral view is currently favored over the invertogram in ARM. A. Puri demonstrated a similar utility of this view in suspected intestinal atresia [12].

The intraoperative suspicion of a CA was raised in our case by findings of a collapsed, fixed descending colon, whereas we expected a dilated descending colon had the pathology only been an ARM. This was noted in two other case reports whereby the microcolon on exploration signified a proximal atresia [7,14]. We reaffirm that the intraoperative findings of a non-distended descending colon in the setting of an ARM have a value of raising suspicion for a proximal obstructing lesion and dictates the need to evaluate the intestinal tract fully.

Management is individualized to the type and level of intestinal atresia, but similarities in approach included staged surgeries with initial resection of the atretic segment, exteriorization via colostomy, or making a vertical enterotomy along the atresia, closed transversely with colostomy [13]. The case series from Japan demonstrated a survival rate of 57.1% [15]. The major factors determining survival were the length of the resected colon and the time to diagnose atresia [2,17]. Delays in referral and limited NICU capacity could have contributed to our post-operative outcome. The limited NICU capacity in the African setting continues to be a significant challenge in achieving satisfactory results in neonatal surgery [18].

The current report emphasizes the importance of an awareness of

possible associated CA with ARM. In our unpublished experience, we have previously encountered a concurrent CA, complete intestinal malrotations and microcolons. We emphasize implementing routine invertograms in a newborn with high ARM and a low threshold for exploration in atypical cases.

4. Patient perspectives

It is very distressing when a newborn is sick, and adequate care requires traveling to another region, whereby they must undergo surgery when they are so fragile. I hope this experience and its documentation improve the outcomes of newborns with this congenital disability.

Provenance and peer review

Not commissioned, externally peer-reviewed.

Sources of funding

No funding was provided for research.

Ethical approval

Muhimbili National Hospital, Institution Review Board Ref. No MNH/IRB-CR/2021/001. Approval for publication granted.

Consent

Written informed consent was obtained from the parents of the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contribution

AI: study conception, production of initial manuscript, collection of data.

L.M, MKN & VN: Study conception, revision of the manuscript, proofreading.

Registration of research studies

1. Name of the registry: RESEARCH REGISTRY
2. Unique identifying number or registration ID: researchregistry6801
3. Hyperlink to your specific registration (must be publicly accessible and will be checked): Browse the Registry - Research Registry

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Declaration of competing interest

No conflicts of interest.

Acknowledgement

We would like to express our gratitutde to the staff of Pediatric Surgery Unit, Muhimbili National hospital. We thank Dr. Sajida, Dr. Wardat A.M., Adellah S. (PhD) and Ella F. T. for their support in prepartion of this manuscript

Appendix A

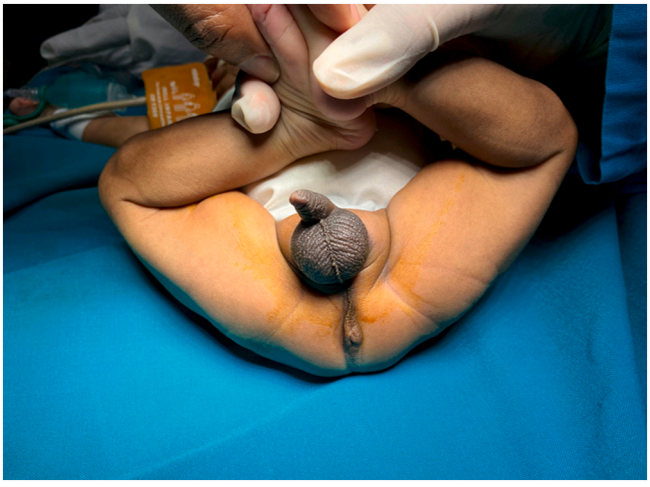

Fig. 1. Perineal examination findings.
Image showing well developed gluteal cleft, anal dimple with no obvious perineal fistula. The external male genitalia and scrotum were also unremarkable.

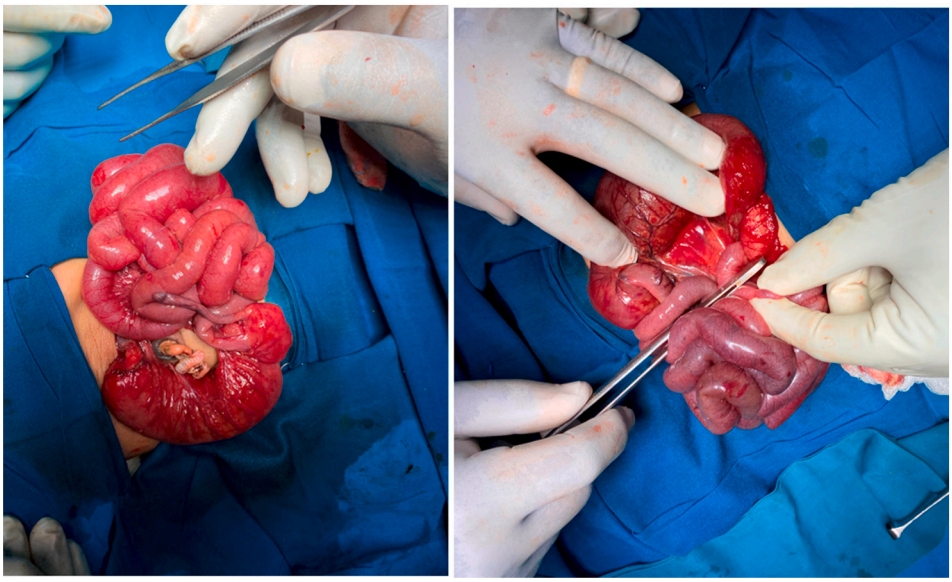

Fig. 2. Intraoperative findings.
Image showing small bowel loops, ceum and appendix, and ascending colon to mid transverse where there is abrupt narrowing, with first type I Bland Sutton and Louw classification A. In image B the other atretic segments is shown distal to the mid transverse one.

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