



## Case report

## Rare case report of anorectal malformation and intestinal atresia



Emily J. Gunderson<sup>a</sup>, Marla A. Sacks<sup>b</sup>, Laura F. Goodman<sup>b</sup>, Asra Hashmi<sup>c</sup>, Andrei Radulescu<sup>b</sup>, Faraz A. Khan<sup>b,\*</sup>

<sup>a</sup> School of Medicine, Loma Linda University, Loma Linda, CA, United States of America

<sup>b</sup> Division of Pediatric Surgery, Department of Surgery, Loma Linda University Medical Center, United States of America

<sup>c</sup> Department of Plastic Surgery, Loma Linda University Medical Center, United States of America

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## ABSTRACT

**Introduction:** Intestinal malformations, such as intestinal atresia, malrotation, and anorectal malformation, are rare events, but concurrent intestinal atresia and anorectal malformation are rare in combination. These anomalies have similar in utero and perinatal symptoms, which can make the diagnosis of both defects prenatally challenging.

**Presentation of case:** We present a case of a male infant with a prenatal diagnosis of truncus arteriosus who on a 32-week routine prenatal ultrasound was suspected to have an intestinal malformation. On day of life one, the patient was taken to the operating room and found to have both type IIIa ileal atresia and anorectal malformation with normal bowel in between. A complete work up for vertebral defects, anorectal malformation, cardiac septal defects, esophageal atresia, renal anomalies, and radial limb defects (VACTERL) anomalies did not reveal additional anomalies.

**Discussion:** In this case, prenatal information supported intestinal obstruction, and the rare combination of both intestinal atresia and anorectal malformation proved surprising and interesting.

**Conclusion:** We suggest providers be aware of the potential of multiple alimentary tract malformations to improve operative preparation and reduce the morbidity or mortality risk from repeat procedures when possible.

## 1. Introduction

Anorectal malformations (ARM) occur in approximately 1 in every 5000 live births while intestinal atresia (IA) incidence is estimated to range between 1.3 and 3.5 per 10,000 live births [1]. The simultaneous presence of both anomalies in one patient is particularly rare [2–8]. The prenatal diagnosis of IA or ARM is possible using ultrasound with a sensitivity of 50% and a specificity of 71% [9]. ARM may be suspected on prenatal imaging, but is usually confirmed on physical exam after birth. Very limited evidence exists to support the use of imaging in prenatal diagnosis of ARM [1]. Early suspicion, management, and surgical treatment of both IA and ARM are necessary to drastically lower mortality and morbidity. This patient, a male infant with suspected ileal atresia on prenatal imaging, was born with an imperforate anus. Later during operative intervention for a divided colostomy was found to have a concomitant ileal atresia. Our case provides an example of the

difficulty in early identification of a concurrent lower intestinal malformation in the neonatal population. This report has been organized in line with the SCARE guidelines [10].

## 2. Presentation of case

The patient is a male neonate conceived through in vitro fertilization and carried as a dichorionic, diamniotic twin by his 28-year-old primigravid mother. Standard prenatal care was followed. The mother had no past medical or surgical history. She denied exposure drug, alcohol, and smoking during pregnancy. Fetal echocardiogram at 32 weeks of gestation identified truncus arteriosus and ultrasound demonstrated large, dilated loops of bowel in twin B. No abnormalities were noted in twin A. Fetal ultrasound at 36 weeks gestation again noted the cardiac abnormalities as well as markedly dilated bowel loops (Fig. 1). At the time, this was considered suspicious for ileal atresia. The mother was

**Abbreviations:** ARM, anorectal malformation; IFALD, intestinal failure associated liver disease; NICU, neonatal intensive care unit; PSARP, posterior sagittal anorectoplasty; VACTERL, vertebral defects, anal atresia, cardiac septal defects, esophageal atresia, renal anomalies, and radial limb defects; TPN, total parental nutrition.

\* Corresponding author at: Loma Linda University Medical Center, 11175 Campus Street, CP21111, Loma Linda, CA 92350, United States of America.

E-mail address: [FAKhan@llu.edu](mailto:FAKhan@llu.edu) (F.A. Khan).

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admitted for induction of labor at 38 weeks gestation at our academic institution, but delivered via Cesarean section after difficulty obtaining fetal heart monitoring on both twins.

Shortly after delivery, the 2550 gram patient was found to have an imperforate anus and admitted to the NICU. APGAR scores were 7 and 9, and 1 and 5 min respectively. At the time of the initial examination by Paediatric Surgery, his vital signs were within normal limits and on physical exam abdomen was soft, non-distended, but significant for imperforate anus without a perineal fistula. A plain radiography of the abdomen (AXR) revealed a dilated loop of bowel suspicious for a dilated sigmoid colon frequently seen in ARM. Nasogastric decompression was instituted while the VACTERL workup was completed, noting the truncus arteriosus.

After thorough discussion, patient and mother agreed to the operation and signed formal consent for the procedure and publication. In the operating room on day of life one, an experienced paediatric surgeon performed a standard left lower quadrant oblique incision for a temporary sigmoid colostomy with mucous fistula to decompress the colon. Upon entering the peritoneal cavity, the child was noted to have a microcolon and the dilated loop of bowel seen on the AXR was related to a Type IIIa ileal atresia just proximal to the terminal ileum. The remainder of the proximal small bowel appeared unremarkable, and later measured 40 cm in length. We decided not to perform a primary tapering enteroplasty and anastomosis at this time due to the inability to adequately interrogate the remainder of the bowel for any additional anomalies and the child's cardiac lesion leading to concern for hemodynamic instability in the operating room with a prolonged operation. An end ileostomy proximal to the atretic segment and a long Hartman pouch were created. Postoperatively, the patient could not tolerate goal feeds due to malabsorption from limited small bowel length, leading to dumping. He began to develop signs of cholestasis from total parenteral nutrition, or intestinal failure associated liver disease (IFALD). A multidisciplinary discussion led to the patient returning to the operating room at 39 days of age by the same paediatric surgeon for re-exploration, ileostomy and mucous fistula take down, ileocolonic anastomosis, and divided sigmoid colostomy, with an aim to improve the absorptive capacity to allow growth.

The infant continued to grow slowly postoperatively and had his cardiac corrective operation at 8 weeks of age. During the initial postoperative period, he was suspected to have a feeding intolerance and the workup was suspicious for anastomotic stricture. The distal colostogram demonstrated a bulbar urethral fistula. The patient returned to the operating room at 13 weeks of age for a revision of the ileo-colic anastomosis stricture and an attempt to perform the posterior sagittal anorectoplasty (PSARP). After extensive adhesiolysis and resection of the original anastomosis, the decision was made to resect and redo the ileocolic anastomosis, leaving the PSARP for a future surgery. The postoperative course was uneventful, but he remained total parental nutrition (TPN) dependent due to dumping, and he was discharged home at 5 months of age. The infant was seen at 6 weeks post discharge in clinic with good wound healing and ostomy management by family. Currently, the patient is being nutritionally supported by a combination of enteral 30% of needs and 70% parenteral routes. PSARP has been planned for 12 months of age.

### 3. Discussion

An infant with anorectal malformation and a concurrent intestinal atresia is an exceedingly rare occurrence. ARM and IA, independently, have the same frequency, occurring in approximately 1 in 5000 live births [11,12]. While VACTERL anomalies are commonly associated with ARM, IA occurs with ARM in every 1 of 246 [1]. The occurrence of both ARM and an intestinal atresia decreases as one moves distally along the intestinal tract. ARM and hindgut abnormalities are less common than concurrent foregut and ARM abnormalities. A review in Japanese literature showed 6 cases of ARM and IA with associated anomalies [13]. ARMs are regularly associated with other malformations with almost every organ system [1,12–14].

Currently, prenatal diagnosis of intestinal atresia occurs most commonly via ultrasound. More distally located intestinal tract abnormalities are more difficult to diagnose because the onset of symptoms indicating intestinal obstruction, such as large bowel dilation and polyhydramnios, are not specific or have a delayed presentation [11]. Ultrasound is less sensitive and specific for these findings when compared



Fig. 1. Multiple dilated peristaltic bowel loops on prenatal ultrasound.

to more proximal atresias [1]. Therefore, while ultrasound is useful for diagnosing intestinal atresia it is less valuable in identifying ARMs. In the most diagnostic cases, it may show dilated colon on screening [13]. More often, ARM is diagnosed with physical exam at birth [1].

Earlier diagnosis, ideally prenatally, has a significant impact on the infant's early hospital course and morbidity. Being able to identify an ARM prior to birth may divert the delivery of the infant to a paediatric surgery center. This will decrease latency of diagnosis at birth at an outside hospital and transportation to a tertiary center. These patients benefit from being at a center with specialists who can diagnosis and promptly treat before a delay which may cause morbidity such as intestinal perforation [15]. Both ARM without perineal fistula and intestinal atresia causing obstruction are urgent surgical indications. The two combined represent an even further increased need for surgical repair. Identifying the incidence of both malformations represents a puzzling diagnostic challenge. Prenatal ultrasound is the currently used diagnostic tool, but it is neither sensitive or specific and the findings present in each malformation overlap significantly.

Knowledge of imperforate anus and intestinal atresia early on, makes a significant impact in patient care. The presence of intestinal atresias, unrecognized prior to going for ARM repair may result in an additional operation. In this case, earlier recognition of the ileal atresia may have led to a different choice in incision site or a laparoscopic, instead of open approach. This may have allowed for fuller access to the atresia and allowed for both repairs during the initial case. Because concurrent intestinal atresia and ARM are regularly associated with further malformations which may also require early surgical repairs, as in our patient, knowledge of both abnormalities is necessary for proper surgical planning. Full knowledge of the defects present would have allowed for more strategic staging of multiple planned surgeries, and ultimately to lower morbidity in our already brittle patient.

#### 4. Conclusion

Although intestinal atresia in association with an anorectal malformation is an uncommon occurrence, practitioners should maintain a reasonable level of suspicion, especially when other fetal abnormalities are noted. Lower intestinal atresias are less common and more difficult to identify. Their preoperative diagnosis is important for guidance of surgical planning and optimal outcomes in affected patients.

#### Consent

Obtained prior to surgery from patient and guardian.

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#### Guarantors

Marla A. Sacks and Faraz A. Khan.

#### CRediT authorship contribution statement

FAK, LFG, AR provided patient care and contributed to manuscript concept and development. MAS contributed to the data collection, analysis, and interpretation. EJG, MAS contributed to writing the manuscript. EJG, MAS, LFG, AR, AH, FAK contributed to reviewing and editing the manuscript.

#### Declaration of competing interest

The authors declare that they have no conflicts of interest.

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