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CASE REPORT

A Triple Whammy of Anorectal Malformation with Malrotation and Jejunal Atresia: A Case Report

Dhruv Jodha Dodiya,[1,*] Sanjeev Agarwal,[2] Dhruv Mahajan,[3] Mit Patel[2] and Sunil Patel[1]

[1]*Resident, Department of General Surgery, Geetanjali Medical College and Hospital, Udaipur*
[2]*Consultant, Department of General Surgery, Geetanjali Medical College and Hospital, Udaipur*
[3]*Consultant, Department of Paediatric Surgery, Geetanjali Medical College and Hospital, Udaipur*

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Abstract

The synchronous association of bowel atresia, intestinal malrotation, and anorectal malformation is rarely seen. Such neonates possess an embryologic, diagnostic, and therapeutic dilemma. Preoperative diagnosis of one in the presence of the other is seldom possible. The trifecta can be confirmed mostly on the operating table only. Thus, the clinician needs to be aware of the possibility of this combination in the presence of any two of these. A full-term neonate presented with the absence of a normal anal opening and multiple episodes of bilious vomiting. On examination, there was upper abdominal fullness and bilious aspirates from the nasogastric tube. A vestibular fistula was identified on perineal examination. The abdominal radiograph had a paucity of distal bowel gas raising a suspicion of proximal bowel atresia. On laparotomy, jejunal atresia type IIIb and malrotation of the midgut were found. Distal atresias were ruled out. A Ladd's procedure and jejuno-jejunostomy were done. The patient had an unremarkable recovery and posterior sagittal ano-rectoplasty was planned at a later stage. Such rarely described anomalies need to be analyzed to get a better understanding of the etiological and embryological basis of such anomalies. It is important to be cognizant of such an anomaly, in case of the defects are seen preoperatively for better planning and outcome.

Keywords: Anorectal Malformation, Malrotation, Intestinal atresia, Bowel atresia

*Corresponding Author: Dhruv Jodha dodiya
Email: dr.dhruvdodiya@gmail.com

Introduction

Neonatal intestinal obstruction can be due to a plethora of causes emanating from some form of developmental abnormality in the gastrointestinal tract embryologically. The association of two separate abnormalities in the same patient has been well described. However, the presence of three separate congenital anomalies in the form of malrotation, jejunal atresia, and anorectal malformation simultaneously is seldom seen with only a few cases reported on the same [1–4]. The coexistence of these possible perilous anomalies poses both diagnostic and therapeutic challenges, as well as requires introspection into the understanding of the embryology of these anomalies. The possibility of synchronous association of these conditions should be kept in mind to avoid intraoperative surprise and to ensure better planning and outcome. We describe a neonate with synchronous jejunal atresia, malrotation of the gut, and vestibular fistula.

Case Report

A three-day-old female child with a birth weight of 1.9 kg born to a 25-year-old primigravida at 39 weeks period of gestation by normal vaginal delivery at home, presented to the casualty with a history of non-passage of stools since birth and bilious vomiting since day two of life. No antenatal scans were done. On examination, the neonate was afebrile, with cold peripheries, tachypnea, and tachycardia. There was mild upper abdomen distension and bilious nasogastric aspirates were obtained. There was an absent anal opening, well-developed gluteal folds, an anal dimple present, and a small perineal fistula just below the fourchette. Normal urethral and vaginal openings were present in the vestibule. Chest auscultation was unremarkable. Laboratory investigations revealed a hemoglobin of 15.8 g/dL, a total leucocyte count of $4.3*10^6$/L, a deranged renal function test (urea- 57mmol/L, creatinine- 1.64mg/dl), and a positive C-reactive protein. However, only mucous output was obtained with no meconium staining. An Abdominal radiograph revealed only a few air-fluid levels in the upper abdomen with a paucity of distal gas. Syringing was tried via the perineal fistula using an 8 Fr infant feeding tube, which could be freely inserted. A possible diagnosis of jejunal or ileal atresia along with perineal fistula was made (Figures 1 and 2).

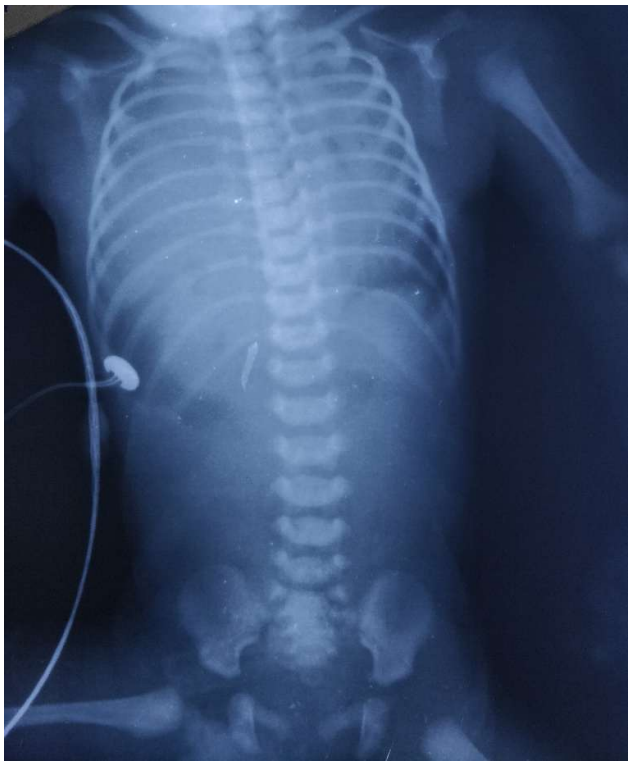

Figure 1. Plain radiograph depicting a paucity of distal bowel gas done at age of 48 hours of life.

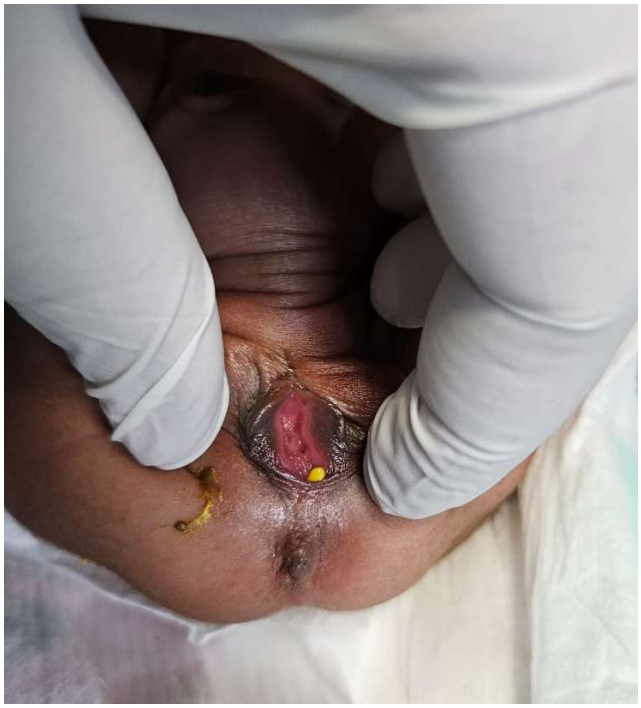

Figure 2: Clinical image of perineum showing vestibular fistula and an absent normal anal opening.

After optimum preoperative optimization and informed consent, the patient was taken up for exploratory laparotomy. Intraoperatively, a jejunal atresia type IIIb (apple peel atresia) with a wide mesenteric defect, and grossly dilated proximal jejunum was found. There was an unexpected associated malrotation of the gut as well. The distal small bowel was completely collapsed with subsequent unused microcolon. Distal atresia was ruled out. Ladd's procedure without appendicectomy and end-to-back jejuno-jejunal anastomosis was done. Post-operatively washes via the fistula and hegar dilation of the fistula were started. The child started passing stools by postoperative day three and nasogastric aspirates decreased gradually. Given clinical suspicion of sepsis along with thrombocytopenia, antibiotics were upgraded; though blood cultures were negative. Slow feeding was instituted from postoperative day five and gradually incremented. The child was discharged on oral feeds with a further plan of posterior sagittal anorectoplasty at a later stage.

Discussion

Congenital anomalies of the gastrointestinal system are one of the more commonly seen congenital anomalies. However, an association of three separate gastrointestinal anomalies in the same patient is comparatively rare. The rarity of this association is explained by the different embryological basis.

Jejunoileal atresia has been attributed to a result of late intrauterine mesenteric vascular occlusions initially by Louw and Barnard[5] and later confirmed in studies on fetal rabbits, dogs, sheep, and chick embryos. Malrotation of the gut, though more commonly seen along with duodenal atresia, can be associated with jejunoileal or colonic atresia as well. The anomalous bowel rotation and fixation leading to midgut volvulus in the intrauterine period can be thought of as the cause of associated type IIIb small bowel atresia.[6] Anorectal malformations, on the other hand, are embryologically related to abnormal hindgut development with impaired septation of the cloaca into the dorsal anorectum and the ventral urogenital tract.[7]

Only a few such cases with a triple association of malrotation, intestinal atresia, and anorectal malformation have been described in the literature. A tabular representation of the individual case findings has been done (Table 1). A variety of associated cardiac, gastrointestinal, and chromosomal anomalies have been described. The correction of malrotation of the gut and a stoma is life-saving for these patients. In case of proximal atresia, duodeno-duodenostomy or jejuno-jejunostomy depending on the atresia location is done. Upfront anoplasty can be done alongside for low anorectal malformations, or definitive repair can be done at a later stage in case of high anomalies. As per the literature review, the prognosis of these patients is satisfactory in most cases.[8] Further studies are required to fully comprehend the embryological basis of synchronous association of these anomalies.

Table 1. A table highlighting the demography, clinical presentation, evaluation, management, and outcome of patients having a combination of malrotation of the gut, intestinal atresia, and anorectal malformation

S. no	Publication	Age at presentation	Sex	Birth details	Birth weight	Local examination	Other Associations	Intraoperative findings	Surgery	Complications	Outcome
1	Ismail et al.	2 days	Male	Full-term, NVD	2600g	Imperforate anus, without perineal fistula with well-formed gluteal cleft and anal dimple	ECHO-normal	Malrotation, type I Bland-Sutton-Louw colonic atresia, ARM	Ladd's procedure, double barrel colostomy	Death	Expired by 72 hours post-operative
2	Kumar et al.	5 days	Female	Full-term, NVD	2100g	Absent anal opening, recently sutured midline perineal wound	ECHO-normal, USG KUB-normal	Malrotation, type IV multiple intestinal atresia	Ladds' procedure, multiple resection anastomosis, high sigmoid colostomy	-	Accepting full orals by 2 weeks post-operatively
3	Morikawa et al.	-	Female	Full-term, Cesarean section	2986g	Anocutaneous fistula	Mild facial characteristics of holoprosencephaly with cleft lip and ocular hypotelorism	Anorectal malformation, malrotation, segmental dilatation of the colon, duodenal stenosis.	Ladd's procedure, endorectal pull-through of the transverse colon proximal to the dilated segment, and cut-back anoplasty	Two months postoperatively- the patient had repeated non-bilious emesis. UGI contrast study and UGI endoscopy- duodenal stenosis,	Underwent duodenoduodenostomy at the age of 8 months. Thriving well at 6 years of age.
4	Morikawa et al.	-	Female	Full-term, NVD	2156g	Imperforate anus	Trisomy 21, cleft palate, large patent ductus arteriosus (PDA)	Duodenal atresia with annular pancreas	Duodenoduodenostomy, Ladd's procedure, and colostomy at the level of the ascending colon	PDA ligation on POD 7. PSARP at the age of 6 months followed by colostomy closure with Nissen fundoplication.	Tolerating enteral feeding through the gastrostomy due to swallowing problems. Bowel movements well controlled with daily enema and good continence.
5	Nitta et al.	1 day	Female	Full-term, NVD	3510g	absent anus apparent anal dimple present. No fistula	-	Membranous atresia of the sigmoid colon	Resection of the atretic colon with a double-barreled sigmoid colostomy	-	Perineal anoplasty at 7 months. Colostomy closure with appendicectomy at 9 months of age.
6	Index case	3 days	Female	Pre-term, NVD	1916g	Absent anal opening, developed gluteal folds, anal dimple present, perineal fistula	SGA	Jejunal atresia type IIIb, Malrotation	Ladd's procedure without appendicectomy, end-to-back jejuno-jejunal anastomosis		

ARM (Anorectal Malformation), ECHO (Echocardiography), NVD (Normal Vaginal delivery), PDA (Patent ductus arteriosus) POD (Postoperative day) PSARP (Posterior sagittal anorectoplasty) SGA (Small for gestational age), UGI (Upper Gastrointestinal), USG KUB (Ultrasound Kidney ureter bladder)

Conclusion

In patients with bowel atresia along with anorectal malformation, a high index of suspicion should be kept for associated malrotation of the gut. Despite the presence of three possibly morbid congenital anomalies simultaneously, a good outcome can be achieved with timely intervention.

Conflict of interest

The authors declare no potential conflicts of interest concerning research, authorship, and/or publication of this article.

Ethics approval

Consent for publication was obtained from parents and the case review was approved by the Institute Ethics Committee.

Consent for publication

Written informed consent was obtained from the patient's parents for the publication of this case report and accompanying images.

Competing interests

The authors declare that they have no competing interests.

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