Clinical Case Reports: Open Access

Yumed Text
OPEN ACCESS JOURNALS

Case Report | Vol 6 Iss 2
ISSN: 2582-5038
https://dx.doi.org/10.46527/2582-5038.257

Diagnostic Dilemma in Teenage Presentation of Intestinal Malrotation: Report of Two Cases and Review of the Literature

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Received: May 09, 2023; **Accepted:** May 25, 2023; **Published:** June 01, 2023

Abstract

Intestinal malrotation is an anomaly of intestinal rotation which occurs during foetal development and usually presents in the neonatal period but may sometimes appear later in life. Its rarity in teenagers portends a diagnostic dilemma for both the surgeon and radiologist hence a high index of suspicion is crucial in diagnosis. We report two cases, a 17-year- old male and an 18-year-old female, who had recurrent episodes of colicky abdominal pain and bilious vomiting due to intestinal malrotation detected at surgery despite doing a preoperative abdominal computed tomography scan (for patient 1) and upper gastrointestinal contrast studies (for patient 2) with neither entertaining a diagnosis of malrotation. These cases emphasize the difficulties and mistakes in diagnosis of intestinal malrotation presenting outside the neonatal period.

Keywords: ***Intestinal malrotation; Teenagers; Diagnostic dilemma; Laparotomy***

1. Introduction

Intestinal malrotation is a clinical entity that encompasses partial to complete failure of the 270 degrees' counter-clockwise rotation of the midgut around the superior mesenteric vessels in the foetal life [1]. Its incidence is one in every 200-500 newborns [2]. The incidence of symptomatic cases is 1 in 6,000 newborns [2]. About 75% to 85% of these patients are diagnosed during infancy, whereas the diagnosis in the rest can be delayed to childhood or even to adulthood [3]. Some individuals actually remain asymptomatic and are never diagnosed throughout their lifetime [4]. At present, the incidence in African children is unknown and there are few reports documenting the occurrence of the condition in older African children [5].

The classical presentation of malrotation in the new born period is bilious vomiting with or without abdominal distension, associated with either duodenal obstructive bands or midgut volvulus [6]. However, the clinical features in the older children or adults may be difficult to elucidate but some available reports have documented recurrent abdominal pain, nausea and vomiting, failure to thrive, abdominal distention, and features of malabsorption syndrome [7,8]. The most consistent finding in older children and adults is the presence of diffuse mild abdominal pain or discomfort which is attributed to the peritoneal bands and this may mimic several other conditions like irritable bowel syndrome, peptic ulcer disease, bilio-pancreatic disease, and psychiatric disorders [9].

Many cases of quiescent malrotation in adults are currently being detected on cross-sectional imaging (particularly computed tomography) for various unrelated reasons [10]. Computed tomography scan does not only show the intestinal malpositioning seen on barium studies but also depicts associated extra intestinal findings not evident on conventional examinations. For example, deviation from the normal relationship between the superior mesenteric artery (SMA) and the superior mesenteric vein (SMV) is a useful indication of malrotation [11].

Occasionally, these cases may be incidentally discovered at laparotomy. In these circumstances it is imperative that adult general surgeons be aware of the embryologic origin and variants of the disorder to manage it effectively [12]. Ladd's procedure is usually performed to correct this congenital defect. This procedure, first described in 1936 by Dr W.E. Ladd begins with counter-clockwise detortion of the bowel, division of Ladd's bands, widening of the small intestine mesentery, and reorientation of the small bowel on the right and the caecum and colon on the left [13].

We aimed at reviewing our experience with these two patients seen over a period of 10 years and to briefly reflecting on this condition as its rarity can lead to serious delay in diagnosis, and unfamiliarity with it may lead to inappropriate surgery.

2. Case Reports

2.1 Case 1

A 17-year-old boy presented to our emergency department with a five-day history of colicky abdominal pain, constipation, and multiple episodes of vomiting. There was no abdominal distension or fever. There was past history of recurrent episodes of upper abdominal and peri-umbilical pain since he was 6 years old and the cause of which remained unidentifiable. There was no history of previous abdominal surgeries or any medical comorbidity.

On examination, he was conscious and coherent, dehydrated with pulse of 84/min and blood pressure of 110/80 mmHg. The respiratory rate was 20/min and temperature was 37.2^{o}C. The abdomen was scaphoid with mild tenderness in the peri-umbilical, epigastric and right hypochondrial regions. His bowel sound was normo-active and rectal examination was unremarkable.

His packed cell volume was 39.7%, white blood cell count was 8000/mm^3, serum potassium was 2.9 mmol/L, urea 56 mg/dL and serum creatinine was 1.2mg/dL. The liver function test result was within normal limits. Abdominal x-ray revealed a large midline significant air fluid level and paucity of gas in the left hemi-abdomen as well as the pelvis suggestive of intestinal obstruction (FIG. 1). There were however no demonstrable dilated loops of bowel. Abdominal ultrasound scan showed

dilatation of the stomach and duodenum, collapsed bowel loops in the right iliac and suprapubic regions with ineffective peristalsis.

He was admitted and managed with nasogastric decompression and intravenous fluid hydration under continuous re-evaluation. Hypokalaemia was corrected. Abdominal computed tomography scan demonstrated a long segment of small bowel with swirling appearance suggestive of one bowel telescoping into another bowel (FIG. 2). There were also demonstrable mesenteric lymph nodes.

The Radiologist suggested a diagnosis of intestinal obstruction from intussusception without evidence of strangulation. He considered mesenteric lymph node as possible lead point.

The patient was worked up for emergency exploratory laparotomy. The findings at surgery were:

- A completely mobilized colon (FIG. 3).
- Dilated stomach.
- Fibrous bands over the distal part of the duodenum and the right side of the abdomen, indicating midgut malrotation (FIG. 4). There was no midgut volvulus.

The congenital bands were carefully dissected and removed, the duodenum was fully mobilized, and the obstruction was relieved. It was then oriented inferiorly towards the right lower quadrant. The mesentery was broadened and the adhesions surrounding the mesenteric vessels were dissected, the entire length of the bowel was examined to ensure that no other obstructive bands were present. After that, the small bowel was placed on the right and the colon on the left sides of the abdominal cavity in a ‘non-rotational position’ (FIG. 5). Finally, the appendix was removed.

He had an uneventful post-operative recovery and was discharged home on the eighth post-operative day. He has been asymptomatic and did well at follow up.

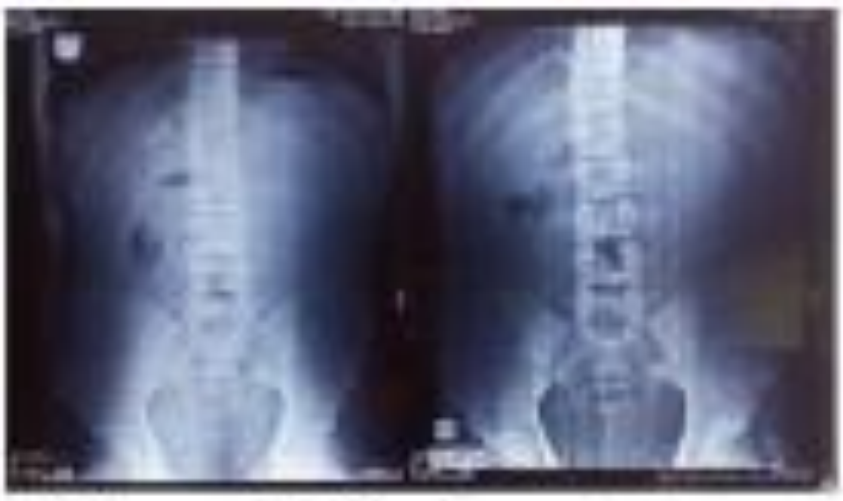

FIG. 1. **Supine and Erect Abdominal X-rays.**

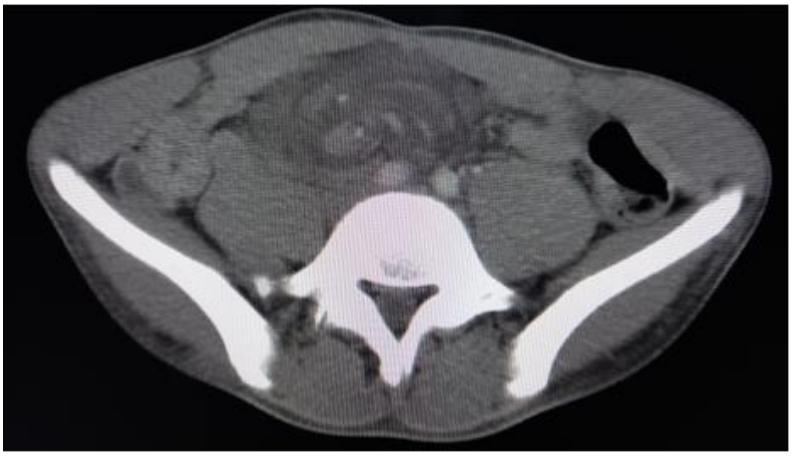

FIG. 2. **CT Scan of the Abdomen.**

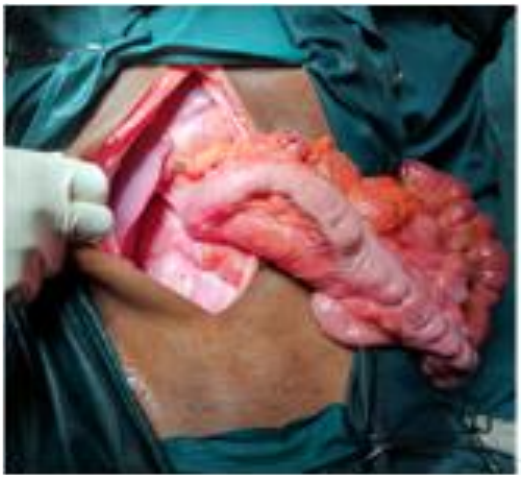

FIG. 3. **Completely mobilized colon.**

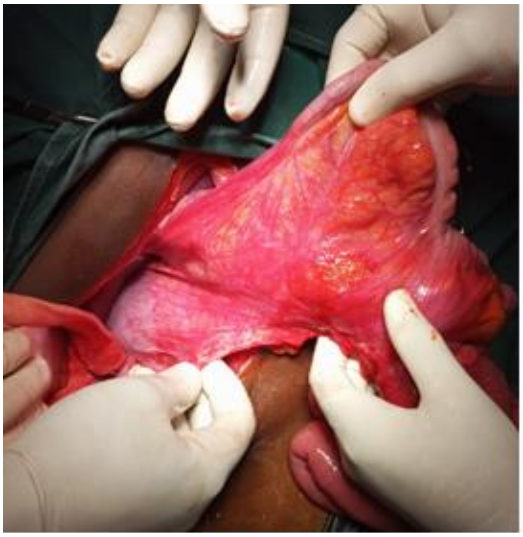

FIG. 4. **Fibrous bands around the duodenum.**

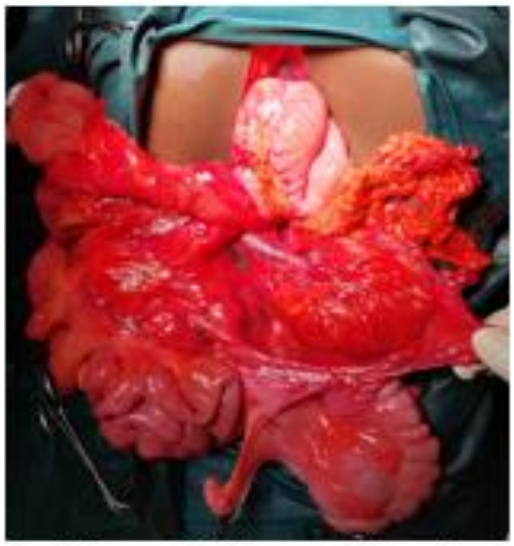

FIG. 5. **Non-rotational placement of the bowel.**

2.2 Case 2

An 18-year-old female undergraduate who has been experiencing recurrent upper abdominal pain and vomiting over the past 8 years presented to the general surgery clinic. The pain was majorly in the epigastric region, colicky, aggravated with meals, non-radiating, non-periodic, no relieving factors, no dyspepsia. Vomiting occurred 2-4 times a day mostly following feeds with each episode containing about 100 ml - 300 ml of bilious effluent on some occasions and at other times, recently ingested food. There was no constipation or abdominal distension and no fever. She had no significant past medical or surgical history. There was associated poor weight gain. Physical examination revealed a young girl who was underweight with epigastric fullness and the rest of the abdomen scaphoid. She had normal bowel sound and rectal examination was unremarkable.

Abdominal ultrasound scan revealed an anomalous vessel in the retroperitoneum around the duodenum and pancreas compressing the duodenum while barium meal showed narrowing of the post bulbar portion of the 2nd part of the duodenum in the prone position only. A diagnosis of small bowel obstruction was considered, and patient counselled for exploratory laparotomy.

Findings during surgery were:

- Freely mobile caecum located in the left paracolic gutter with an uninflamed appendix measuring about 10 cm (FIG. 6).
- Collapsed small intestine disposed to the right paracolic area and abnormally located duodenojejunal junction.
- Dilated duodenum, up to the 3rd part, with anomalous vessel crossing the anterior aspect of that part, compressing adherently on the duodenum with a kink (FIG. 7).
- External Ladd's band holding the duodenum to the retroperitoneum.
- Other organs appeared normal and in their usual anatomic position.

An intra-operative diagnosis of malrotation was made. Release of Ladd's bands and abnormal vessel causing duodenal obstruction was done with the mesentery broadened as well as appendicectomy. The bowel was then placed in the position of non-rotation.

The patient had a good recovery and was discharged on post-operative day 6. She maintained a progressive improvement with weight gain on subsequent follow-up visits.

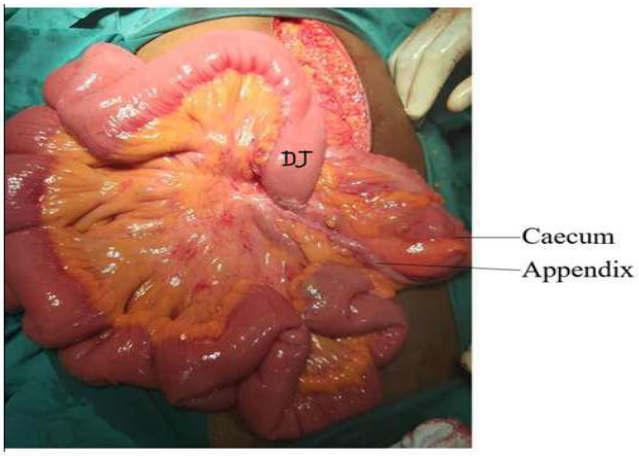

FIG. 6. **Freely mobile caecum and appendix in the left paracolic area close to the duodenojejunal (DJ) junction.**

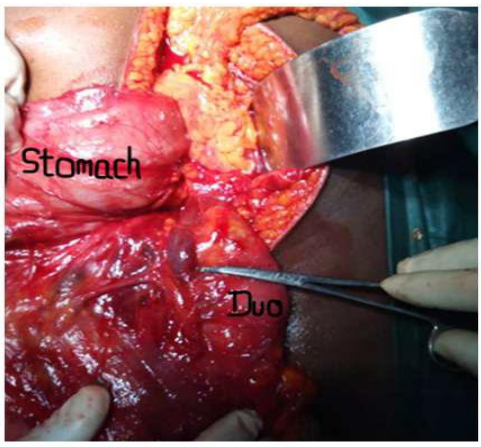

FIG. 7. **Haemostat pointing at the anomalous vessel crossing the anterior part of the duodenum.**

3. Discussion

Malrotation is a congenital anatomical anomaly of foetal intestinal rotation. It is defined by abnormal position of the bowel within the peritoneal cavity and is usually accompanied by abnormal mesenteric bands or absence of fixation of portions of the bowel [4,14]. There is a spectrum of appearances depending on the embryological stage of development at which the anomaly occurs, ranging from omphalocoele to non-rotation, incomplete or reversed rotation, internal hernia and mobile caecum/ unattached duodenum [15]. Malrotation can lead to the life threatening complication of midgut volvulus, which can result in obstruction, strangulation and necrosis of the entire midgut [14].

Presentation of intestinal malrotation is very rare with an incidence between 0.2% and 0.5% [16]. Approximately 85% of malrotation cases present in the first two weeks of life but that in older children and adult is unclear [17]. The clinical diagnosis in adolescents and adults is difficult because it is rarely considered on clinical grounds. The difficulty of diagnosis results from both the absence of specific physical findings and the low frequency in older children or adults [18]. In our case reports, both diagnoses were made intra-operatively.

Symptoms of intestinal malrotation are linked to the anatomical anomalies associated with it [19]. Two distinct patterns of presentation in older children and adults have been reported in the literature: acute and chronic [20]. The latter is more common. This is characterised by intermittent crampy abdominal pain, bloating, nausea and vomiting over several months or years. The symptoms may be highly non-specific. However, the range of clinical presentations underlines the need for a high index of suspicion of midgut malrotation as the cause of intermittent and varying abdominal symptomatology in a healthy young adult [20]. Diagnostic delays are common in this group of patients because of the non-specific nature of the presentations. The pathophysiology of these chronic symptoms may relate to the compression effect of Ladd's bands running from the caecum and ascending colon to the right abdominal wall [20]. The patients with acute presentation usually present with features of acute bowel obstruction with or without chronicity of symptoms. In the older child or adult with malrotation, midgut volvulus is the most common cause of bowel obstruction [21].

Our index patients appeared to have been asymptomatic in childhood but started developing symptoms from early adolescence which progressed over time. In reviewing the literature on the patterns of presentation, Dietz et al reported that 5 adults out of a series of 10 with bowel obstruction caused by malrotation studied presented with chronic features and that the duration of symptoms even extended to 30 years [22]. Fu et al. [23] reported that 6 of 12 patients in their series presented with chronic intermittent abdominal symptoms. Adeniyi et al reported a case series of 5 older children with gut malrotation who presented to the Lagos University Teaching Hospital (LUTH) in Southwest Nigeria with recurrent abdominal pain and/ or vomiting between January 2013 and October 2015 [5]. In a case series of gut malrotation by Nasir et al. [24] in 2011, 9 cases of gut malrotation were documented and 2 of the cases were aged 3 and 13 years respectively. These children also presented with recurrent abdominal pain [24]. Nwankwo and Gboobo in Port Harcourt observed essentially the same symptomatology in the older children audit of patients with gut malrotation over a 5-year period [25]. Kotobi et al. [26] noted that majority of patients in their series had chronic presentations with misdiagnosis for a long time and inadequate surgeries when they did, resulting in recurrence of symptoms. Izes et al and Haak et al also reported that patients had chronic presentations although the patient reported by Haak et al had features of acute intestinal obstruction at presentation [27,28] just like our first case report who had episodic abdominal pain for a period of eleven years but presented with features of acute duodenal obstruction. Fernandez-Moure et al. [29], however, in a case report described an older patient with acute presentation and no history suggestive of chronicity. Similar presentations were also reported by singh et al and Emanuwa et al [30,31].

Diagnostic features of midgut malrotation can be identified using plain abdominal radiograph, ultrasound scan (USS), computed tomography (CT) scan, magnetic resonance imaging (MRI) scan and mesenteric arteriography [14]. Conventional plain radiography is neither sensitive nor specific in the diagnosis of gut malrotation, although right sided jejunal markings and the absence of a stool-filled colon in the right lower quadrant may be suggestive, leading to further investigation. Abdominal

colour Doppler USS may reveal malposition of the SMA, raising the suspicion of gut malrotation with or without the abnormal location of the hollow viscus [14].

The reported gold standard for the diagnosis of gut malrotation is an upper gastrointestinal (UGI) contrast study, particularly in the paediatric age group [14,20]. It's accuracy is over 80% and the rules familiar to paediatric radiology also apply for adults-that is, the duodenal-jejunal junction fails to cross the midline and lies below the level of the duodenal bulb [32]. Contrast enema examination usually shows malposition of the right colon with the ileum entering the caecum from the right, but the caecum may assume a normal location in up to 20% of patients. This normal location may cause malrotation to be missed on this type of study [33]. Contrast findings therefore may be non-specific and a normal study does not exclude the possibility of gut malrotation [14,20]. This may be the case in the second patient reported above where the upper GI contrast study done failed to diagnose the malrotation.

Computed tomography scan of the abdomen with contrast study is increasingly used preferentially as it is now considered as the investigation of choice, providing diagnostic accuracy of 80% [14,20]. CT and MRI scans may show the SMV to be in an anomalous position, posterior and to the left of the SMA. In addition, they may show the abnormal anatomical arrangements of the midgut with the duodenum not crossing the spine. Deviation from the normal positional relationship of SMV and SMA was originally described by Nichols and Li as a useful indicator of the diagnosis of midgut malrotation [34]. However, abnormal orientation of the SMA-SMV relationship is not entirely diagnostic of malrotation; it can be seen in some patients without the pathology and a proportion of patients with malrotation may have a normal SMA-SMV relationship [14]. The CT appearance of midgut volvulus is diagnostic of malrotation. The shortened mesentery allows the small bowel and mesentery to twist and wrap around the narrowed SMA pedicle to create a distinctive 'whirlpool' appearance on CT scan. This pattern was first described by Fisher in a patient with midgut volvulus [35]. It can be detected with both abdominal USS and CT scan. Our 2 cases, however, did not have volvulus.

Mesenteric angiography was previously used but is now rarely indicated in the evaluation of malrotation. Angiography was used to demonstrate the characteristic corkscrew appearance of a whirling SMA and its branches; the 'barber pole sign' as well as extensive collaterals caused by proximal SMA occlusion [36]. However, its role has been superseded by the CT scan which has the overall advantage of not only detecting the abnormal location of the midgut but also the reversed mesenteric anatomical relationship as well as any other intra-abdominal anomalies associated with malrotation. The CT scan done for our first patient failed to report classic findings of malrotation. An important learning point from this case is that CT imaging may not provide a conclusive diagnosis in complex cases such as ours but can be helpful for localising a pathology and planning for surgery even though the diagnosis may not have been accurate from the CT scan.

The management of intestinal malrotation was first described by William Ladd for paediatric patients in his 1936 landmark surgical paper and this hasn't changed much since then except for the laparoscopic approach that has been described by Van der Zee and currently carried out by surgeons skilled in this approach [37,38]. In general, symptomatic patients should be treated with surgical intervention. Though, Spigland et al. recommended that all patients with malrotation are candidates for laparotomy, even if they are asymptomatic, because the complications associated with intestinal malrotation are based on anatomic reasons that do not alter with age [39]. The classic treatment is the Ladd's procedure which requires mobilization of

the right colon and caecum by division of Ladd's bands, mobilization of the duodenum, division of adhesions around the SMA to broaden the mesenteric base, and an appendicectomy to prevent future diagnostic dilemma of an abnormally located appendix [40]. We offered our patients this procedure. In addition, we separated the abnormal vessel causing duodenal compression in our second patient. Of note is that one needs to keep in mind that the general surgeon may not necessarily be as familiar with the procedure when compared to the paediatric surgeon so a paediatric surgeon's assistance may be required. The decision to carry out the procedure with the paediatric surgeons in our 2 patients seems appropriate when viewed retrospectively considering how uneventful their post-operative periods were and the fact that our patients were completely symptom free during our 12 months of follow up.

4. Conclusion

Intestinal malrotation, a rare condition in the teenage patient portends a diagnostic dilemma for both the surgeon and radiologist in the clinical setting. The difficulty of diagnosis results from both the absence of specific physical findings and the low frequency of the condition. Increased awareness of this condition and an understanding of its varied presentation at different ages may reduce the time needed to diagnose the problem. Early diagnosis and immediate operative intervention are key factors associated with a better patient outcome.

5. Conflict of Interest

None declared.

6. Author Contributions

All authors participated in the evaluation and/or treatment of the patients, critical review and revision of the manuscript and gave final approval for submission.

7. Ethical Statement

The authors certify that written informed consent was obtained from the patients' parents for publication of these reports and any accompanying images. The patients understand that their names and initials will not be published, and due effort will be made to conceal their identities.

8. Acknowledgement

The authors thank the two patients for the acceptance to publish their cases.

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