The radiological finding of chronic malrotation in a 12-year-old child with omphalocele and liver herniation: a case report and literature review

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European Journal of Medical Case Reports
Volume 5(8):246–248
https://doi.org/10.24911/ejmcr/173-1620750924

ABSTRACT

Background: Malrotation of the intestines is an uncommon pediatric condition that typically presents in the first month of life.

Case Presentation: Using the SCARE 2020 Guidelines, we report a case of a 12-year-old female with omphalocele and bilobed liver herniation presenting with chronic intestinal malrotation. A computed tomography of the abdomen and pelvis with IV contrast revealed a peritoneal defect measuring 4 cm in the upper abdomen, containing the gut including pylorus, duodenum while retrospective distension of the stomach was seen up to the pylorus. The open Ladd's procedure was performed to correct the defect. No complications were noted intra- or 1 month post-operatively. Patients with intestinal malrotation may present with abdominal pain, intestinal obstruction, nausea, or vomiting. Conclusion: The open Ladd's procedure was deemed safe, feasible, and effective in a resource limited country like Pakistan, in the treatment of young children with intestinal malrotation.

Keywords: Intestinal malrotation, omphalocele, liver herniation, Ladd's procedure, Pakistan, case report.

Received: 11 May 2021 **Accepted:** 05 August 2021 **Type of Article:** CASE REPORT **Specialty:** Radiology

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Background

Malrotation of the intestines is not an uncommon pediatric condition that typically presents in the first month of life. It is a clinical entity that comprises of partial or complete failure of the 270°C clockwise rotation of the midgut around the superior mesenteric vessels during the fetal life [1]. Symptomatic presentations are associated with 1 per 6,000 live births, where around 90% of the cases of intestinal malrotation present within the first year of life [1]. Intestinal malrotation presenting in adults is an extremely rare phenomenon that accounts for up to 0.5% of total cases [2]. The presentation of malrotation secondary to congenital anomalies in a 12-year-old child is a rare occurrence. Using the SCARE 2020 guidelines [3], we report a case of a 12-year-old female with omphalocele and bilobed liver herniation presenting with chronic intestinal malrotation.

Case Presentation

A 12-year-old female presented on 21st October 2020 to the emergency department with a lump in the epigastric region with sudden "thumping" pain, which was graded 9 out of 10. The legal caretaker reported that the pain commenced 8 hours earlier when the patient started crying holding her stomach. The patient reported no dysuria, rectal bleeding, fever or any change in bowel habits. Overall, her health as well as the past medical and surgical history was unremarkable.

On examination her respiratory rate was 32 breaths per minute and pulse rate was 90 beats per minute and regular; all other vital signs were within normal range. The patient had marked epigastric tenderness, involuntary guarding and mild abdominal distension. Her laboratory investigations showed, hemoglobin 11.5 g/dl, white blood cells 7.6 × 103/ µl, aplatelets 358 × 103 /µl and Erythrocyte sedimentation rate (ESR) 15 mm during the first hour. Blood urea, creatinine, and electrolytes were all within normal range. The abdominal ultrasound showed normal liver, gall bladder, pancreas, kidneys, and urinary bladder, while the epigastric lump appeared as a solid structure measuring 12 × 10 cm (largest dimension) with parenchyma similar to liver. The color flow Doppler (ultrasound) showed blood vessels within the lump. The patient was treated symptomatically. A computed tomography (CT) of the abdomen and pelvis with intravenous contrast revealed a 4 cm long peritoneal defect in the upper abdomen, containing the pylorus and the duodenum with upstream gastric stasis (Figure 1a,1b). Liver was bilobed with one lobe in the herniated sac subcutaneously measuring 12.3 cm

(largest dimension) along with gall bladder, thus appearing as gastrochisis. The second hepatic lob measuring 11 cm was in the right hypocondrium without focal lesion or intrahepatic biliary dilatation. The portal vein and its tributaries were normal. Jejunum loops were present in right hemi-abdomen. Inverse orientation of superior mesenteric vein to superior mesenteric artery (SMA) was diagnostic of malrotated gut. There were visible slices through the base of the lungs that were unremarkable.

The patient was prepared for surgical correction of the defect using open Ladd's procedure at the District Headquarter Hospital, Gujranwala, Pakistan, which also gave the ethical approval for publication of this case report. Written informed consent was taken from the guardian. There were no complications intra- or one-month post-operatively.

Discussion

Omphalocele is a congenital anterior abdominal wall defect with an umbilical herniation of intestine covered by a layered membrane of peritoneum, Wharton's jelly and amnion. There has been a frequent association of omphalocele with other congenital anomalies [4]. During embryogenesis at week 6, the midgut undergoes herniation through the umbilicus, which is physiological. During that time, intestinal elongation and counterclockwise rotation around the SMA occurs. The midgut undergoes spontaneous reduction around 10 to 12 weeks back into the abdominal cavity. An omphalocele occurs when the lateral folds do not meet in the midline. It infrequently presents alone and is usually associated with other anomalies such as pentalogy of Cantrell, Beckwidth-Wiedemann syndrome, as well as anal, cardiac or gut malrotation. There is also an association with maternal age with development of omphalocele [5].

According to the Stringer classification, the malrotation has been classified as type-1 non-rotation, type-2 duodenal malrotation and type-3 duodenal plus caecal malrotation [6]. In this case, there was type-2 duodenal malrotation, which is linked to the 6th and 10th week of intrauterine life, wherein the gut herniates into the umbilical cord and only involves the small bowel.

It is frequently found to be associated with Gastrointestinal tract (GIT) malformations (duodenal atresia stenosis or web), biliary system malformations (agenesis of the gall bladder intra- and extra-hepatic biliary atresia), pancreatic malformations (hypoplasia or agenesis of the dorsal pancreas), congenital diaphragmatic herniation, heterotaxy, choanal atresia, and hypospadias [7]. Abnormalities like these are often found incidentally on CT when some other condition is being investigated, but regardless diagnosis is almost always delayed and often even missed [8].

Intestinal malrotation etiology is unclear. It has been suggested that the forkhead box transcription factor and L-R asymmetry genes as causative factors, while autosomal dominant and recessive inheritance patterns have also been linked [8]. Malformations such as Martinez-Frias syndrome present with malrotation as well as multiple gastrointestinal tract atresias and abnormalities of the pancreas plus biliary system [7]. It has also be linked to chromosomal abnormalities such as trilogy of the long arm of chromosome 16 and a ring chromosome 4 [9]. Clinical malrotation presentation varies greatly with age, with common presentation in infants as mid gut volvulus, as compared to only 15% in the adult group [10]. Common symptoms are bloating, weight loss, nausea, vomiting, and abdominal pain.

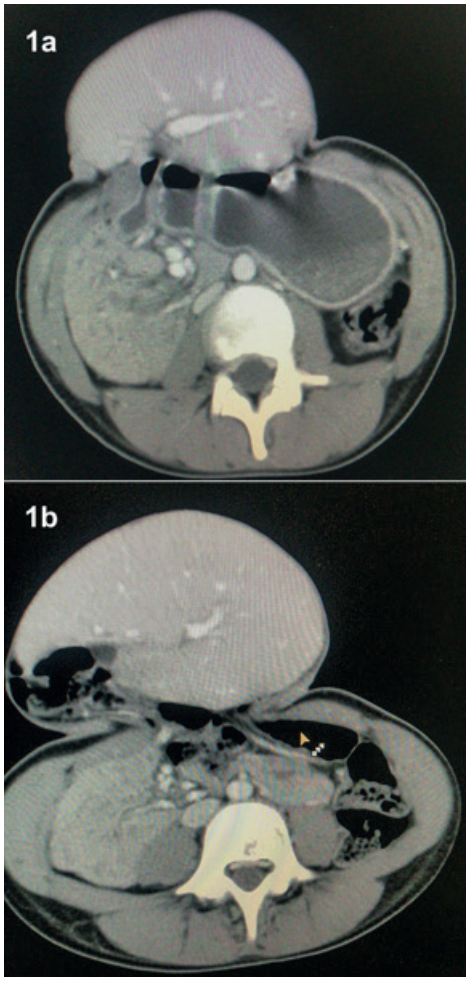

Figure 1. *(a and b). CT scan abdomen and pelvis (with contrast). Malrotated gut: the findings are likely gastroschisis with bilobed liver; one lobe is in the herniated sac subcutaneously and the other lobe is in the abdominal cavity.*

Conclusion

While asymptomatic adults may not require surgery, patients with acute volvulus would likely require prompt treatment. Increased awareness of this condition and its presentation in developing countries like Pakistan may reduce the time to diagnosis and improve patient outcomes.

What is new?

Malrotation of the intestines is an uncommon pediatric condition that typically presents in the first month of life and is a rare occurrence as the age advances. Using the SCARE 2018 criteria, we report a case of a 12-year-old female with omphalocele and bilobed liver herniation presenting with chronic intestinal malrotation. The open Ladd's procedure was performed, and considered safe, feasible, and effective in a resource-limited country like Pakistan, in the treatment of young children with intestinal malrotation.

List of Abbreviations

CT	Computed tomography
ESR	Erythrocyte sedimentation rate
GIT	Gastrointestinal tract
SCARE	Surgical case report
SMA	Superior mesenteric artery

Conflict of interests

All authors declare that there is no conflict of interest regarding the publication of this Case Report.

Funding

No funding was obtained.

Consent for publication

Written consent was obtained from the patient (parents of the patient).

Ethical approval

Ethical approval was not required at our institution to publish an anonymous case report.

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Summary of the case

1	**Patient (gender, age)**	Female, 12
2	**Final diagnosis**	Malrotated gut with omphalocele and liver herniation
3	**Symptoms**	Lump in the epigastric region with sudden "thumping" pain, graded 9 out of 10
4	**Medications**	No relevant medical history
5	**Clinical procedure**	Surgical correction of the defect using open Ladd's procedure
6	**Specialty**	Radiology, surgery