Adult Intestinal Malrotation: A Diagnostic Challenge - A Case Report and Literature Review

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Abstract

Adult intestinal malrotation is a rare anatomical anomaly that typically manifests during infancy but can also present in adulthood. Symptoms are mainly digestive, with a long history of intermittent abdominal pain and epigastralgia. This often presents a diagnostic challenge due to the varied and nonspecific nature of its clinical symptoms. Radiological evaluations revealed diverse patterns of malrotation, including incomplete rotation, mesenteric base abnormalities, and associated malformations. CT scans studies consistently identified characteristic anatomical distortions, aiding in accurate diagnosis.

In this context, we present a unique case in which contrast-enhanced computed tomography (CT) of the abdomen, initially conducted to investigate a suspected episode of acute pancreatitis accompanied by epigastralgia, fortuitously revealed the presence of intestinal malrotation.

Once the diagnosis has been made, the therapeutic approach is based on monitoring or managing complications such as intestinal obstruction.

Early recognition and accurate radiological assessment play a pivotal role in establishing the diagnosis and guiding appropriate management strategies. Increased awareness among clinicians and radiologists is crucial to prevent diagnostic delays and potential complications associated with adult intestinal malrotation.

Categories: Gastroenterology, Radiology, General Surgery

Keywords: small / abnormalities*, intestine, computed tomography, pancreas / abnormalities*, congenital abnormalities, adult intestinal malrotation

Introduction

The intestinal malrotation is a congenital developmental anomaly of the digestive tract during foetal life. During the complex process of this development, errors in rotation of the umbilical loop can sometimes give a serious complication [1].

With an incidence of 1 per 6,000 births, this anomaly is diagnosed during the first year of life in 90% of cases, but some cases remain asymptomatic [2].

Extremely rare in adults, with an incidence of between 0.00001% and 0.19% [1], most intestinal malrotations are diagnosed incidentally during surgery or imaging [3,4].

In adults, symptoms are principally characterized by sporadic abdominal pain, epigastralgia, biliopancreatic manifestations, and vomiting, although a subset of patients remains asymptomatic [5]. The correlation with congenital anomalies, especially those pertaining to the pancreas, is typically fortuitously identified [6-8].

The gravest complication pertains to the occurrence of complete small bowel volvulus, a phenomenon observed in approximately 1 to 2% of the instances delineated within the realm of paediatric cases [9,10].

We report a rare case of a 32-year-old male patient presenting with acute abdominal pain of pancreatic type who was incidentally found by CT to have a complete common mesentery with hypoplasia of the uncinate process of the pancreas.

Case Presentation

The 32-year-old male patient presented for consultation due to an episode of acute transfixing abdominal
pain, which was accompanied by radiating discomfort to the back, epigastralgia, and concurrent nausea. Notably, the patient also exhibited respiratory symptoms characterized by a productive cough yielding greenish sputum.

Pertinent aspects of the patient’s social history included ongoing tobacco use with a cumulative exposure of 10 pack-years, sporadic alcohol consumption, and persistent occupational stress. Notably, there was an absence of any prior surgical interventions or documented drug allergies in the patient’s medical history.

The patient remained hemodynamically stable, exhibiting a blood pressure of 115/68 mmHg, a pulse rate of 72 beats per minute, and an oxygen saturation level of 98% while breathing room air. Abdominal examination yielded no notable abnormalities. Auscultation unveiled a few deviations tied to the symptoms of cough and secretions, while the individual’s respiratory status remained unaltered. The prominent presenting symptom entails acute, intense pancreatic pain accompanied by epigastralgia, which has manifested over a span of 48 hours.

Following these assessments, an abdominal-pelvic CT scan, performed with contrast injection, was conducted with the aim of detecting signs of acute pancreatitis. The findings disclosed a configuration in which the entire length of the small bowel, inclusive of the duodenojejunal juncture, assumes a position to the right of the spinal column, while the entire colon aligns to the left (Fig 1a & Fig 1b).

![CT scan images](image_url)

**Figure 1:** The CT scan shows, in axial (a) and coronal (b) sections, a colon on the left side of the abdomen (white star), and the small intestine on the right (arrow head).

Notably, there is an absence of a third portion of the duodenum within the aorto-mesenteric clamp setup (Fig 2a). Additionally, there is an inversion in the customary anatomical relationship of the superior mesenteric vessels, as evidenced by the vein traversing to the left of the artery (Fig 2b).
FIGURE 2: CT images demonstrating (a) the absence of the 3rd portion of the duodenum in the aorto-mesenteric clamp and (b) the abnormal superior mesenteric artery-vein (SMA-SMV) relationship associated with intestinal malrotation; With the SMV (arrow head) being to the left of SMA (full arrow).

Coincidentally, a diagnosis of uncinate process aplasia of the pancreas has been incidentally ascertained; this condition exhibits no irregularities concerning the pancreatic ducts (Fig 3a & Fig 3b). The ultimate diagnosis remains indicative of a complete common mesentery, concomitant with uncinate process aplasia of the pancreas, albeit devoid of any involvement of pancreatic parenchyma.

FIGURE 3: Axial section (a) of an aplastic uncinate process. No pancreatic tissue is visible behind the SMV. (b) For clarification, the pancreas with its globular head is marked in blue, highlighting the aplasia of the uncinate process.

It is imperative to acknowledge that the quality of CT images falls short of being optimal for a comprehensive exploration of the digestive tract. The utilization of digestive endoscopy was omitted in the assessment of the gastric mucosa’s surface. Within this context, the absence of any abnormalities pertaining to the pancreatic parenchyma, peri-pancreatic fat, or aberrations in the pathological uptake of contrast agents is noteworthy.

Investigations have revealed a normal electrocardiogram (ECG) and negative troponin levels, measured at 0.0001 ng/ml. Lipasemia levels were recorded at 21.7 IU/l (reference range: 8-78), while hyperleukocytosis was noted at 12200/mm3. The haemoglobin concentration stood at 14.5 g/dl, accompanied by a platelet count of 422 x 10^3/L. Both creatinine and blood urea nitrogen (BUN) values fell within acceptable ranges. Lipid profiles exhibited moderate disturbances, notably with suboptimal levels of high-density lipoprotein (HDL) at 0.37 g/L, whereas serology for Helicobacter pylori (HP) yielded negative results. This moderate hyperleukocytosis could potentially be linked to a bronchopulmonary infection.
Through clinico-biological investigation, the presence of a myocardial infarction in the inferior region has been ruled out via scrutiny of electrocardiogram (ECG) results and troponin levels. The primary concern lies in the potential occurrence of a small bowel volvulus, a condition frequently unveiled through the identification of this anomaly. Our therapeutic approach encompassed administering symptomatic relief and maintaining vigilant surveillance over the patient to pre-empt any complications.

Discussion

Intestinal malrotation is a congenital developmental anomaly. It is a rare entity, occurring in 1 in 5000 cases, with 90% of cases diagnosed in the first year of life [2,11,12]. Other reports publish a higher incidence of 1 in 500 births [1,12-14]. The increase in incidence may demonstrate the importance of advances in imaging detection, although the validity of these incidences remains questionable. This study considers only non-rotation and no other types of intestinal malrotation [14]. The incidence of this anomaly in adults is estimated between 0.00001% and 0.19% [1], and Autopsy diagnosis revealed population figures around 0.03% [15].

This anomaly is frequently associated with congenital malformations and morbidity, making it essential to evoke this diagnosis at an early stage by a CT scan with contrast injection, to optimize patient management [1,9]. In adults, the symptoms of malrotation are non-specific and multifaceted [2,4,11].

Dominated by abdominal pain, with 76.8% in a review of the literature as reported by Neville and al., constipation, nausea, vomiting and bowel obstruction may be observed [3,5,15]. Rarely, pancreatic symptoms are described in the literature with 2.1% [7,16]. The abnormal anatomical relationship between the duodenum and pancreas may be responsible for intermittent obstruction of the pancreatic duct [7].

The physiopathology of malrotation has not been identified, but some authors such as Martin and al. have identified a genetic factor, demonstrated by the association of malrotation with mutations in the forkhead box transcription factor (FOXF1) and L-R asymmetry genes [9,11,14]. Other factors include consanguinity, recessive or dominant transmission, chromosomal imbalance, and environmental factors. In other conditions, intestinal malrotation is a syndrome of an associated underlying pathology, such as Martinez-Frias syndrome [6,14].

The embryogenesis of the primitive intestine is complex, and in 1817, Meckel was the first to describe a physiological hernia of the primitive intestine. In 1898, Mall explained in his publications the reintegration of this primitive intestine into the abdominal cavity by counterclockwise rotations and fixations at 270° on the SMA axis. In 1915, Frazer and Robbins presented the three stages of intestinal rotation, still used today two centuries later [9,13]. The first stage, the primitive loop, is located outside the abdominal cavity. It gradually undergoes an initial 90° counterclockwise rotation centered on the SMA. The duodenojejunal angle is to the right of the SMA, and the ileo-caecal junction to its left [4,9]. At 10 weeks, the second stage develops, which consists mainly in the integration of the primitive loop into the abdominal cavity through a second 90° counterclockwise rotation, which completes a 180° rotation from the initial position. At this stage, the ileo-caecal junction is positioned in the subhepatic region, the duodenojejunal angle remains to the right of the SMA, and the first and last ileal loops are next to each other [4,9,13]. At 11-12 weeks, the third stage of rotation develops, consisting of a 90° counterclockwise rotation of the primitive loop (i.e., a 270° rotation from the initial position) into the abdominal cavity [6,9,13]. At the finals, a duodenojejunal angle is found below the SMA, ending up to the left of the spine. The ileo-caecal junction passes in front of the SMA and is positioned in the right flank. The first jejunal loop then passes into the left hypochondrium and the last ileal loop into the right iliac fossa [5,9,13].

The abnormal rotation is the premature interruption of rotation at the different stages described previously. In this way, Stringer proposed a classification into three types of these anomalies by the stage at which the embryonic rotation error developed: type 1 (no rotation), type 2 (duodenal malrotation) and type III (combined duodenal and caecal malrotation) [12,17,18].

Congenital anomalies are very common in intestinal malrotation, accounting for up to 60% [6]. Thus, a defect in intestinal rotation during embryogenesis closely involves the duodenum and consequently disrupts the development of the pancreas [7,8]. The majority of the literature describes hypoplasia or aplasia in the process of unciform development [2,9,18]. Inoue and al. reported aplasia or hypoplasia of the uncinate process in a series of five cases with malrotation, and normal uncinate process in 101 patients without malrotation [8]. Chandra and al. reported 86% abnormal development of the head of the pancreas in a series of 25 cases of non-rotating bowel, 86% of which had aplasia or hypoplasia [8]. This malformation may be associated with an anomaly of the pancreatic ducts, exposing patients to the risk of Wirsung canal plication and causing a defect in drainage of the pancreas [7,9].

The inversion of the relationship of the superior mesenteric vessels, where the artery passes to the right and behind the vein, is another essential feature described in intestinal malrotation [1,2,18,19]. Chandra and al., in a series of 25 cases, found 90% of cases with this anomalous position of the mesenteric vessels [8], and in another systematic review of 194 cases, Neville and al. found 58% inversion of the SMA/SMV ratio [16]. The
vessels’ position is an essential feature observed on cross-sectional imaging but is not diagnostic [16].

The imaging modality is an exemplary diagnostic tool, and contrast-enhanced computed tomography (CT) continues to maintain its gold-standard status [11,15,20], with a sensitivity rate of 97.5%, as reported in the systematic review by Neville and al. [12,16]. Meticolous assessment of the entire bowel, combined with clear differentiation of the superior mesenteric artery and vein, not only informs the diagnostic process but also improves our understanding of diagnostic findings [5,20]. Ultrasound, conversely, is an appropriate imaging option for paediatric patients and infants, with a sensitivity rate of 62.5%. However, it lacks a substantial clinical indication for the diagnosis of malrotation [2,15,16], conventional radiography with opacification and MRI have a lower yield.

This case is a non-rotation (type 1), which include a left positioned cecum and ascending colon with a duodenojejunal junction to the right of the rachis, accompanied by an inversion position of the superior mesenteric vessels and aplasia of the uncinate process of the pancreas. The essential role of imaging, especially CT scans, is particularly emphasized in this context, and this work encourages radiologists to be extremely familiar with this pathology to predict the ultimate complications, which remain small bowel volvulus or small bowel obstruction. In incomplete common mesentery, these complications are more frequent, because of the reduction in length of the root of the mesentery and are reported in several publications [9-11,18].

Conclusions
This case highlights a rare presentation of malrotation, in an adult, with aplasia of the acinar process of the pancreas, which manifested as acute abdominal pain, raising suspicion of pancreatitis. It is rarely described, in the literature, that intestinal malrotation can manifest as pain suggesting pancreatitis, and many publications describe that this congenital anomaly was discovered incidentally during surgery for conditions other than intestinal malrotation, such as appendectomy. However, these patients have previously benefited from radiological examinations such as CT scans and are sometimes referred back to surgery. Therefore, we emphasize the importance of raising awareness among practitioners and, especially, radiologists, so that early detection and accurate radiological assessment play an essential role in diagnosing this pathology, guiding appropriate therapeutic strategies and ensuring that potential complications associated with intestinal malrotation are not missed.

Additional Information
Disclosures
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