ISSN 2689-8268 | Volume 7

American Journal of Surgery and Clinical Case Reports

Case Report Open Access

Polysplenic Heterotaxy Syndrome (Left Isomerism) – Case Report and Consideration for the Surgical Treatment. A Rare Case of Occluding Colon Cancer in a Patient with Polysplenia and Intestinal Malrotation

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Received: 26 Feb 2024

Accepted: 06 Apr 2024

Published: 12 Apr 2024

J Short Name: AJSCCR

Keywords:

Heterotaxy Syndrome; Left Isomerism; Polysplenia; Intestinal malrotation; case report

Citation:

Vorburger SA. Polysplenic Heterotaxy Syndrome (Left Isomerism) – Case Report and Consideration for the Surgical Treatment. A Rare Case of Occluding Colon Cancer in a Patient with Polysplenia and Intestinal Malrotationt. Ame J Surg Clin Case Rep. 2024; 7(13): 1-4

1. Abstract

Heterotaxy syndrome is a rare situs anomaly. In contrast to situs inversus, not all visceral organs are mirrored. We present a case featuring left isomerism heterotaxy syndrome, characterized by polysplenia and the left-sided positioning of the colon, in a patient diagnosed with stenosing adenocarcinoma in the proximal colon. A 47-year-old woman presented with a 5-month history of postprandial cramps, distended abdomen and weight loss. Investigations identified a highly stenotic adenocarcinoma in the proximal colon. The CT revealed bilobed lungs, right-sided polysplenia, a left-sided colon, and a midline position of the liver. The intestinal malrotation affected the surgical strategy for the oncologic tumor resection due to the vascular and lymphatic anomalies. The preoperative detection of left isomerism allowed optimal preparation for surgery with CT angiography of the abdominal and iliac arteries. Due to the folding of the colon in the left hemiabdomen, the mesocolon presented with 4 layers on top of each other. Intraoperative findings showed that the tumor invaded several layers of this mesocolon. We performed laparoscopic adhesiolysis, extended proximal hemicolectomy en-bloc with several mesocolonic layers, and a side-to-side ileo-midcolonic anastomosis. The postoperative course was uneventful, and the patient left the hospital after 6 days. Histological findings confirmed a R0 resection of the T4 tumor. To our knowledge, this rare coincidence of left isomerism and occluding adenocarcinoma of the colon has never been described. Preoperative planning of the surgical strategy with 3D CT scan and CT angiography enabled an oncologic sound operation despite the uncommon anomaly.

2. Introduction

2.1. Definition and Classification of Heterotaxy Syndrome

Unlike situs inversus, where all organs are mirrored, situs ambiguous, or heterotaxy syndrome, also features non-mirrored organ positioning. This genetic disorder results in anomalous positioning of organs and blood vessels relative to the midline. Conversely, situs solitus describes the normal organ arrangement with the heart, spleen, stomach, and aorta on the left, and the liver and vena cava on the right [1]. Situs anomalies are categorized into situs inversus and situs ambiguous. Situs inversus is the mirror image positioning of organs, whereas situs ambiguous lacks proper lateralization, leading to symmetrical arrangements (isomerism) in paired structures like the lungs and midline positioning with potential malformations in unpaired abdominal organs [2,3]. Situs ambiguous is further divided into cases with polysplenia and cases with asplenia, or right and left isomerism, respectively (Figure 1).

Isomerism defines a situation in which morphologically right or left structures are found on both sides of the body. Right isomerism features bilateral trilobed lungs, two right atrial cavities, often with asplenia, leading to immune deficiencies, early cyanotic heart disease diagnosis, and respiratory distress. Due to these factors, right isomerism is commonly identified during early childhood, often resulting in premature mortality. Conversely, left isomerism, less severe in heart disease, may go undiagnosed into adulthood, char-

acterized by bilateral bilobed lungs, two left-morphology atriums, and typically presents with polysplenia and an interrupted inferior vena cava [1]. We report a case involving a 47-year-old female with left isomerism heterotaxy syndrome.

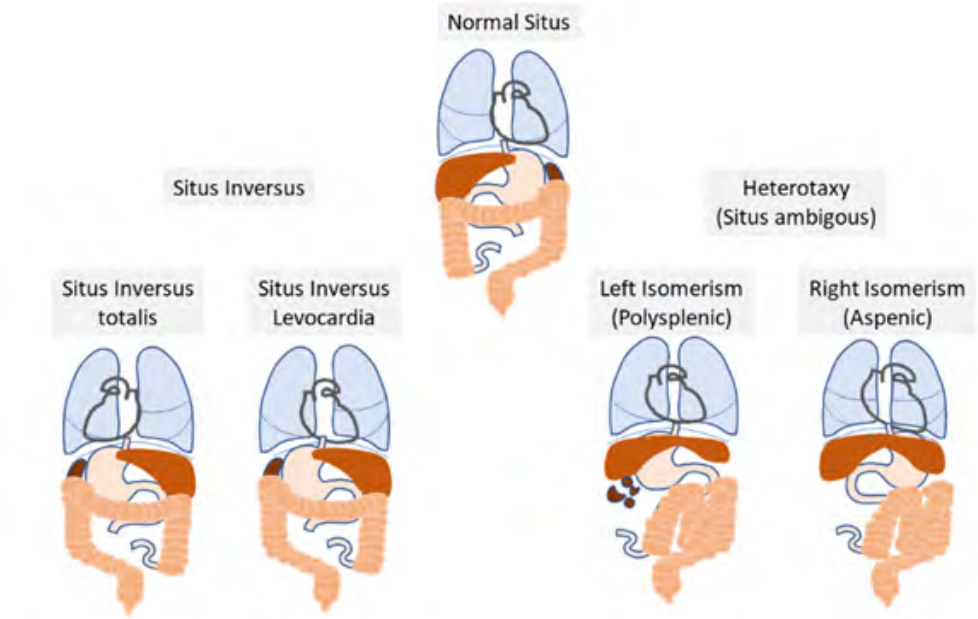

Figure 1: Classifications of situs anomalies. The figure shows the anatomical dispositions of the viscera in different types of situs anomalies.

3. Case Presentation

We report the case of a 47-year-old female patient, primipara, with no history of prior intraabdominal interventions. The patient had a known history of uterine myoma and cholecystolithiasis. She presented with cramping abdominal pain, nausea, and emesis. Over the past five months, the patient experienced recurrent postprandial cramps accompanied by a distended abdomen and malaise. Additionally, she noted an unintended weight loss of 5 kg recently. Prior to this episode, the patient was asymptomatic. Physical examination revealed a distended abdomen with palpable resistance and tenderness in the left hemiabdomen. Further investigations via CT scan and colonoscopy identified a stenosing adenocarcinoma in the proximal colon without evidence of distant metastasis. Additionally, the CT scan revealed intestinal malrotation with left positioning of the colon, bilobed lungs bilaterally, and polysplenia (Figure 2 and 3), findings that are compatible with left isomerism heterotaxy syndrome.

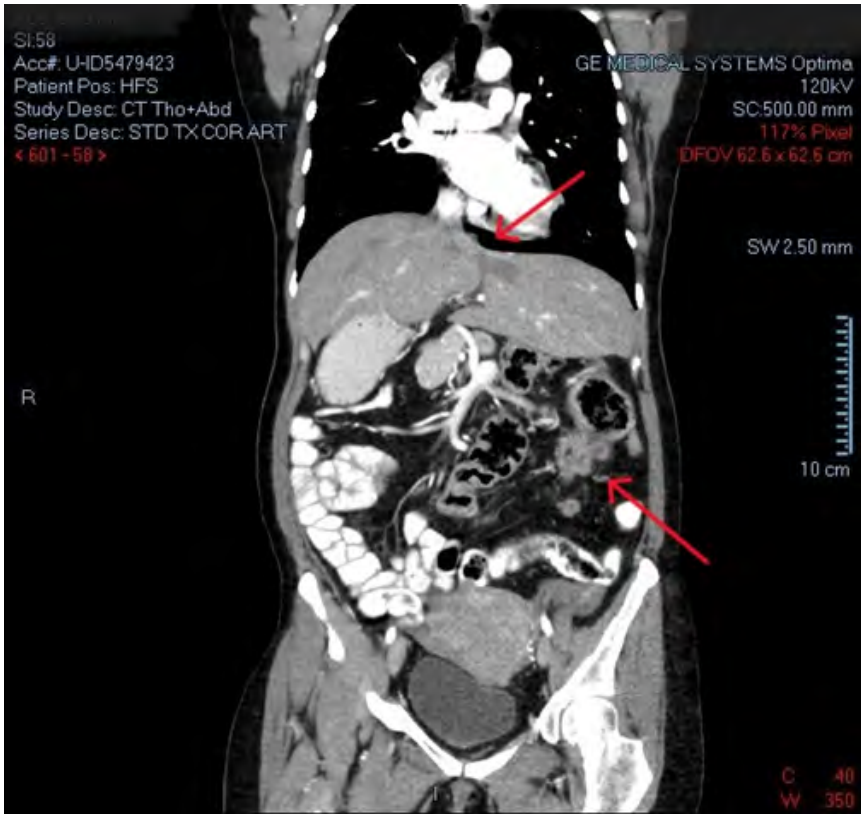

Figure 2: Tumor in the proximal colon and midline position of the liver on abdominal CT scan (arrows).

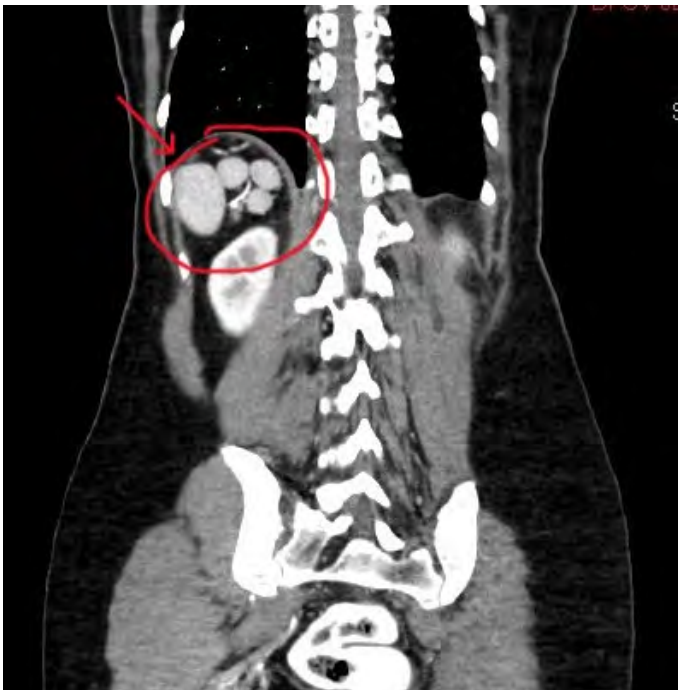

Figure 3: Polysplenia: Multiple, right-sided spleens on abdominal CT scan (circle).

4. Perioperative Findings

The laparoscopy showed a polysplenic heterotaxy with incomplete intestinal rotation. We found turbid ascites in the small pelvis and alterations on the uterus and left ovary. Both the small intestine and proximal colon were dilated, necessitating initial decompression to facilitate surgery with an adequate visual field. The colon was folded over itself several times before it drained into an almost normal left flexure: Prestenotically, we found an ascending and a descending part, poststenotically another three ascending and - with the colon descendens - three descending intestinal parts, all located in the left hemiabdomen. Consequently, there were four overlapping layers of the mesocolon. The tumor had infiltrated the mesocolonic layers posteriorly, involving the "true" colon descendens and the "colon transversum", which lay behind the "right" hemicolon (Figure 4a and 4b). The release of adhesions and mobilization of the "right" hemicolon and "transverse" colon took approximately 120 minutes before resection could commence.

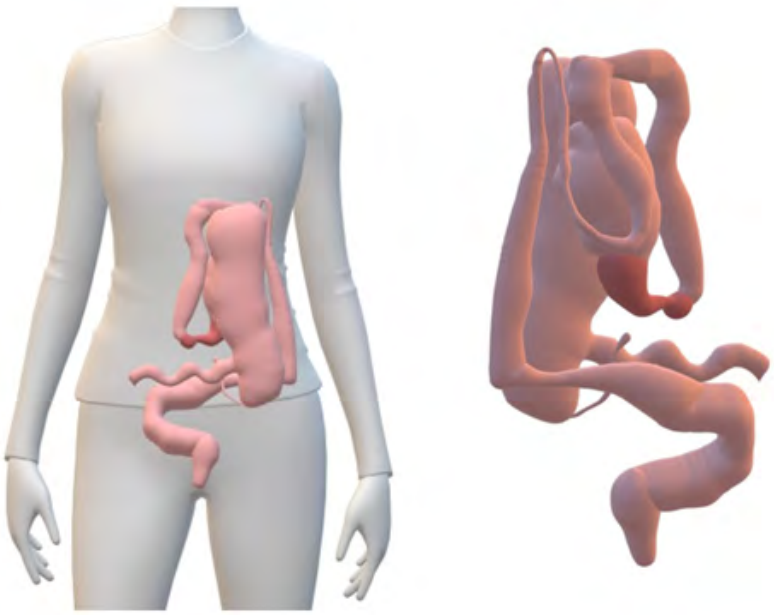

Figure 4a and 4b: 3D Reconstruction of colon in situ. 3D reconstruction model of the left colon with stenosing tumor in the proximal colon. a) frontal view b) view from dorsal

5. Postoperative Course

We discharged the patient on the sixth postoperative day in a good general condition. Histological analysis confirmed the adenocarcinoma, which was classified according to the TNM Classification (UICC, 8th Edition, 2016) as pT4a, pN1b (2/49), cM0, R0. Subsequently, the patient underwent adjuvant chemotherapy. The 12-month follow-up revealed no evidence of tumor recurrence.

6. Discussion

Heterotaxy syndrome is a rare finding, with an estimated incidence of 1 per 10,000 to 20,000 live births [1]. The etiology of heterotaxy remains incompletely understood. However, it is recognized that over 80 genes play a role in normal right-left organ development, including ZIC3, ACVR2B, NODAL CRYPTIC, CRELD-1, NKX2.5, SHROOM 3, and LEFTY 2. Patterns of autosomal dominant, autosomal recessive, and X-linked inheritance have been documented [1]. Because intestinal malrotations typically remain asymptomatic in adults, their true incidence is challenging to ascertain. Often, such anomalies are only identified incidentally in the context of unrelated diseases, as in the present case. Given the scarcity of documented cases in medical literature, reporting these cases is important [4–9]. The diagnosis of left isomerism heterotaxy syndrome in this patient was made via CT scan, initially performed to evaluate colon cancer. Considering the potential for atypical blood supply in individuals with heterotaxy syndrome, we conducted a preoperative 3D CT scan and CT angiography to delineate arterial anatomy accurately, thereby reducing the risk of postoperative intestinal ischemia. Identifying the heterotaxy syndrome preoperatively enabled thorough surgical planning, facilitating a minimally invasive approach and effective oncological resection with central lymph node resection while preserving arterial perfusion to the remaining colon.

7. Conclusion

Understanding heterotaxy syndrome, including its range of vascular and visceral abnormalities, is essential for the effective management of these rare conditions. Preoperative planning with the aid of 3D CT scans and CT angiography facilitates a comprehensive oncologic resection, even in the face of unusual anatomical variations.

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