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Case Report

Surgical Management of Obstructive Biliary Disease in a Patient with Situs Ambiguous: A Case Report

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ABSTRACT: Situs Ambiguous is a rare congenital disorder, which varies in the degree of disease severity. It poses a high rate of mortality when it is associated with congenital heart disease, immunodeficiency, or intestinal obstruction; otherwise, it may only present with minor symptoms or none at all. Despite modern-day advancements in medical technology, the detection of this disease is limited to radiologic imaging, most often warranted when investigating for another disease entity. This report highlights the case of a 49-year-old Filipino female, diagnosed with obstructive jaundice secondary to a choledocholithiasis. An endoscopic retrograde cholangio-pancreatography (ERCP) was performed for biliary clearance but was unsuccessful due to an alleged ampullary mass. However, while imaging proved this ampullary mass to be non-existent, she was incidentally identified to have situs ambiguous, which could account for the failed initial intervention. The patient underwent open cholecystectomy with intraoperative cholangiography, and common bile duct exploration via choledochoscopy, which the patient tolerated well.
Keywords: *choledocholithiasis; intraoperative cholangiography; open cholecystectomy; situs ambiguous*

INTRODUCTION

Situs ambiguous is an extremely rare syndrome with an overall incidence of 4 per 1 million live births.[1] Also known as heterotaxy syndrome, this condition is defined as an abnormal pattern of anatomical organization of thoracoabdominal organs, involving the heart, lungs, liver, spleen, stomach, and bowels.[2] This is also associated with a disrupted specific turn pattern during embryologic development.[3] Heterotaxy can be divided into two subtypes; [1] right (atrial) isomerism, which is associated with asplenia and a higher mortality rate due to associated congenital heart disease, immunodeficiency, and intestinal obstruction[4] and [2] left (atrial) isomerism, which is associated with polysplenia and a significantly lower mortality rate.[5] Situs anomalies do not directly cause symptoms in patients; however, the uncertainty of organ location confuses clinicians when patients are managed for surgical conditions, such as appendicitis or cholecystitis.[6] We present a case report of an adult Filipino female patient with the polysplenic form of situs ambiguous, who presented with obstructive jaundice secondary to choledocholithiasis, but had an unsuccessful biliary clearance by endoscopic retrograde cholangio-pancreatography due to an atypical location of the ampulla of Vater.

CASE

A 49-year-old female presented with a three-week history of epigastric pain and jaundice, associated with postprandial vomiting and tea-colored urine. The pain was described to be colicky in character, radiating to the back, and aggravated by heavy meals. On physical examination, the patient was jaundiced, with icteric sclerae and direct tenderness on the epigastric area.

Ultrasound showed contracted gallbladder with a thickened wall, but no evident intraluminal lithiasis noted. Intrahepatic ducts and proximal common bile duct (1.46cm) were dilated, with a 1.38cm lithiasis noted within the common bile duct. The right liver lobe was contracted with an AP diameter of 8.61cm, and the left liver lobe was prominent with an AP diameter of 10.97cm with slightly wavy contour, suggestive of liver cirrhosis. Apart from the unusual hepatic dimensions, there was no mention of any aberrancy in terms of the organs' locations. CBC analysis showed WBC at 11.40 x 109/L (reference: 4.5-10 x 109/L). Total bilirubin levels were elevated at 12.79mg/dL (reference: 0.20–1.19mg/dL), with the direct fraction measuring 9.69mg/dL (reference: 0.00-0.50mg/dL), and indirect fraction measuring 3.15mg/dL (reference: 0.20-0.70mg/dL). Alkaline phosphatase was also found to be elevated at 291 U/L (reference: 40-150 U/L). Alanine Aminotransferase (ALT) was also elevated at 378 U/L (reference: 0.00-55 U/L). Findings pointed to biliary obstruction secondary to a choledocholithiasis, which warranted an endoscopic retrograde cholangio-pancreatography (ERCP). ERCP was unsuccessful due to a severely deformed D1-D2 junction, attributed to either an ampullary mass or a bulging ampulla.

A CT scan of the whole abdomen with IV and oral contrast was done to investigate further, and demonstrated the anomalies associated with situs ambiguous (polysplenia type) -- the liver was located midline, with the left liver lobe appearing larger compared to the right with smooth contour. The gallbladder was also located midline and noted to be partially contracted with no calcified lithiases observed within its lumen. The intrahepatic, common hepatic (1.6cm), and common bile duct (1.2cm) were dilated with a cluster of faint densities measuring 0.5 x

0.3cm noted at the distal common bile duct. Also, within the distal common bile duct was a punctate calcific density measuring 0.3 x 0.2 x 0.2cm. The pancreas was malpositioned with its head located anteriorly and left of midline, while its body and tail coursed posteriorly and to the right side. The stomach was located on the right side of the upper hemiabdomen inferior to the liver, with the duodenum crossing the midline anteriorly to the left side (Figures 1 and 2). The small bowels were on the left side of the hemiabdomen, while the large bowels were on the right, with no signs of bowel obstruction (Figure 3). Multiple splenules were visualized in the right upper hemiabdomen, posterior to the stomach, and lateral to the right kidney (Figures 4 and 5). An important revelation in the CT scan was the absence of the ampullary mass, which was previously suspected after an unsuccessful ERCP.

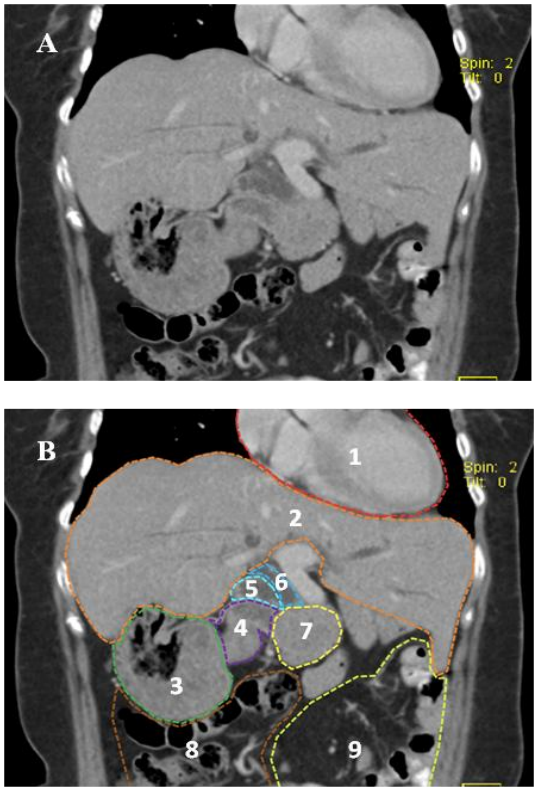

Figure 1. A. CT scan of whole abdomen with IV and oral contrast (coronal section). **B.** Coronal section labeled as follows: (1) heart with normal configuration, (2) liver at the midline, (3) stomach on the right hemiabdomen, (4) duodenum crossing the midline anteriorly to the left side, (5) partially contracted gallbladder at the midline, (6) dilated extrahepatic ducts, (7) pancreas with head located anteriorly and left of the midline, (8) large bowels on the right hemiabdomen, (9) small bowels on the left hemiabdomen.

Following unsuccessful biliary clearance through ERCP, the patient was then advised to undergo open cholecystectomy with intraoperative cholangiography and common bile duct exploration due to the anticipated difficulty of a laparoscopic surgery in the setting of situs ambiguous. A midline incision was done for adequate exposure. The liver, described in the CT scan as having a smooth contour, was noted to be grossly cirrhotic intraoperatively. Otherwise, the locations of the visualized viscera proved to be consistent with the CT scan findings. The liver was enlarged, with the left lobe extending to the left hemiabdomen. The contracted gallbladder was at the midline and was isolated via dome down technique. Intraoperative cholangiogram through the gallbladder fundus defined the biliary anatomy, demonstrating the location of the cystic duct and bile ducts. The intraoperative cholangiogram revealed dilated right and left intrahepatic ducts with a cut off at the proximal common bile duct (Figure 6). Common bile duct exploration was completed with a 1cm stone extracted using Randalls forceps through a transcystic approach from the middle third of the common bile duct. Biliary clearance was established using a choledochoscope and completion cholangiogram (Figure 7). The ampulla was likewise visualized on choledochoscopy; and no mass was appreciated. The patient tolerated the procedure well with an unremarkable post-operative course, and was discharged three days post-surgery.

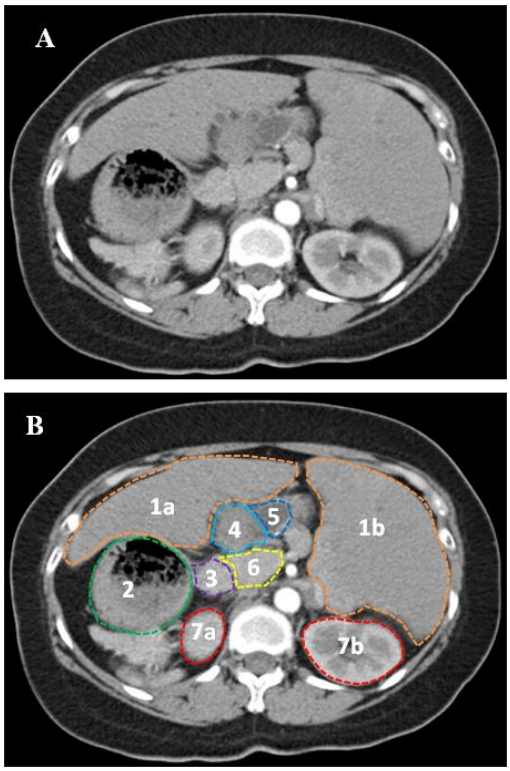

Figure 2. A. CT scan of the whole abdomen with IV and oral contrast (axial section). **B.** Axial cut labeled as follows: (1a) right liver lobe, (1b) left liver lobe, (2) stomach, (3) duodenum, (4) gallbladder, (5) extrahepatic duct, (6) part of the pancreas, (7a) right kidney, (7b) left kidney.

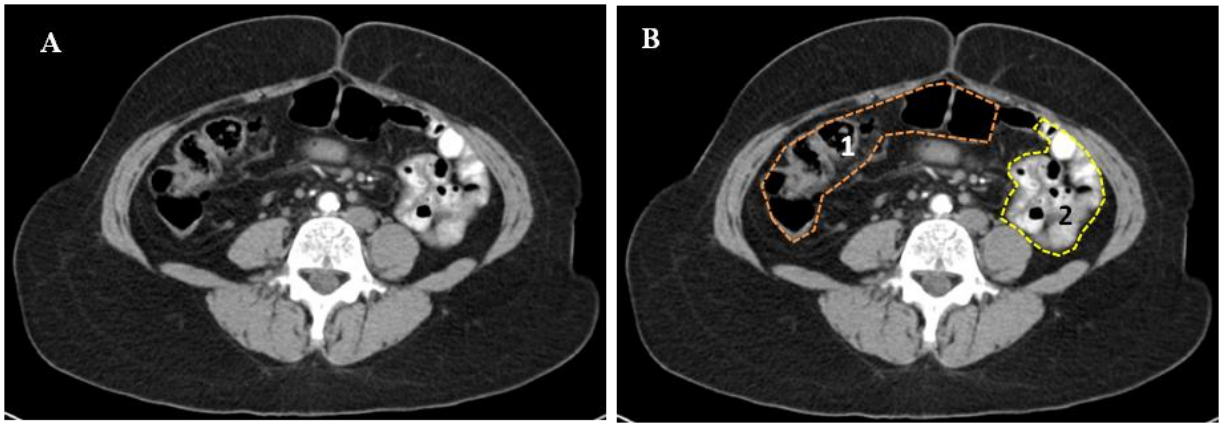

Figure 3. A. CT scan of the whole abdomen with IV and oral contrast (axial section). **B.** Axial cut labeled as follows: (1) large bowels, (2) small bowels

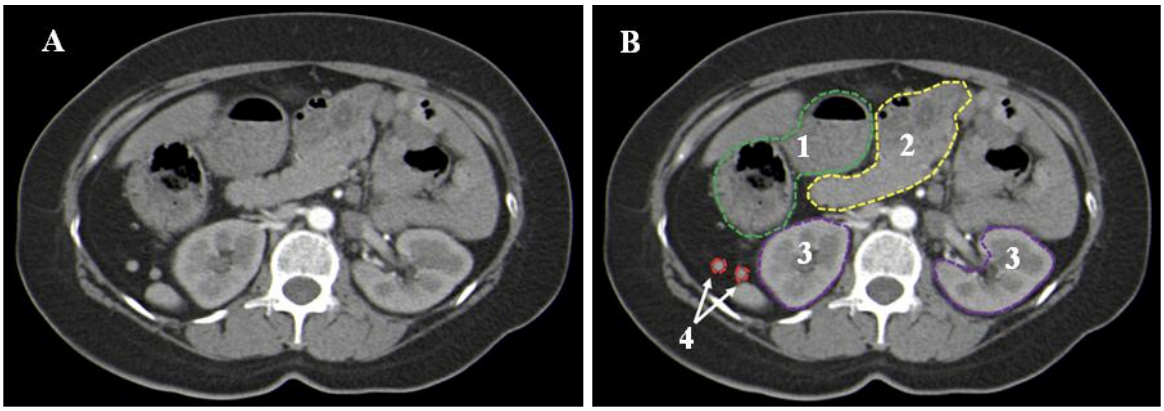

Figure 4. A. CT scan of the whole abdomen with IV and oral contrast (axial section). **B.** Axial cut labeled as follows: (1) stomach, (2) pancreas, (3) kidneys, (4) splenules on the right hemiabdomen, posterior to the stomach and lateral to the right kidney.

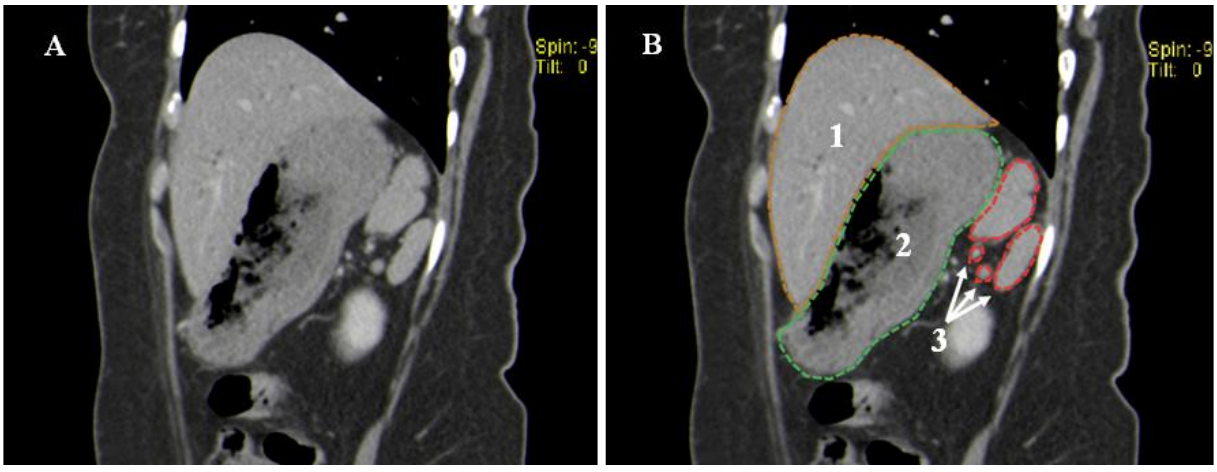

Figure 5. A. CT scan of the whole abdomen with IV and oral contrast (sagittal section). **B.** Sagittal section labeled as follows: (1) liver, (2) stomach, (3) spleen with multiple splenules.

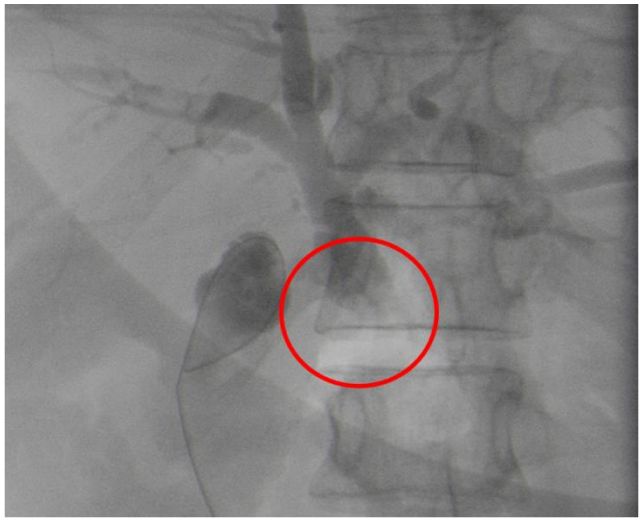

Figure 6. Intraoperative cholangiogram showing dilated right and left intrahepatic ducts with cut-off at the proximal common bile duct.

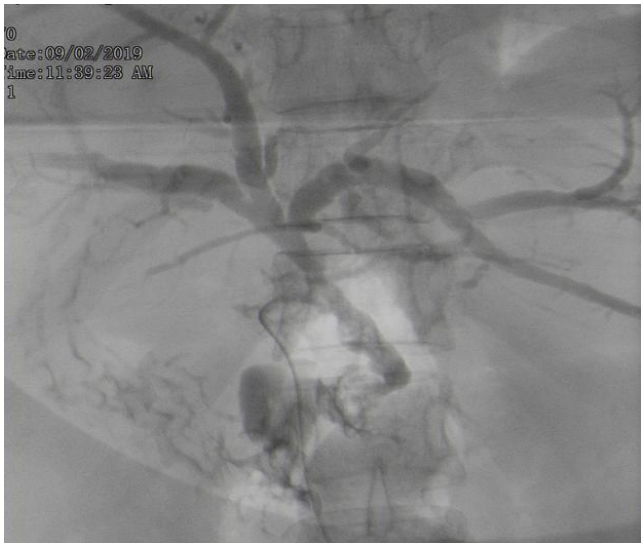

Figure 7. Completion cholangiogram after extraction of choledocholith showing egress of contrast into the duodenum signifying biliary clearance.

DISCUSSION

Situs ambiguous presents as an abnormality in the left-right axis, consequently resulting in an anomalous arrangement of heart chambers, lungs, and abdominal organs. This usually results in loss of structures (e.g. asplenia), gain of structures (e.g. polysplenia), and/or failure to regress certain symmetric embryonic structures (e.g. persistent left superior vena cava).[7] Its pathophysiology is rooted in mutations or chromosomal imbalances that affect the following: ZIC3, CFC1, ACVR2B, LEFTY2, NKX2.5, CRELD1, NODAL, CRIPTO, and GDF1, thereby altering the early developmental processes responsible for embryonic left-right axis determination.[8] Multiple modes of inheritance have been described, from autosomal recessive to X-linked recessive to autosomal dominant with incomplete penetrance.[9] Recent data show maternal environmental influences in addition to genetic predisposition and chromosomal mutations as the cause of the anomalous defects.[10] The prevalence of situs ambiguous, including both right and left atrial isomerism forms, is estimated at 0.8 per 10,000 individuals.[10] Situs ambiguous has two subtypes that often overlap: left atrial isomerism with a polysplenic association and right atrial isomerism with an asplenic association.[11]

Except for the cardiac and pulmonary abnormalities, this case is characteristic of the classic and usual arrangement of abdominal organs in situs ambiguous. There was found to be polysplenia, described as multiple splenules visualized in the right upper hemiabdomen, posterior to the stomach and lateral to the right kidney. This finding classifies this patient under the polysplenia type of situs ambiguous. The liver, gallbladder, and biliary tract are located at midline, which explains the patient's symptom of epigastric pain.[3] Moreover, the stomach is mirrored, hence located on the right upper hemiabdomen. Bowels may typically have varying degrees of malrotation, which in this case were arranged as follows: the small bowels on the left side, while the large bowels were on the right side of the abdomen.

Our patient is unique since she did not have any cardiac abnormalities as shown in her electrocardiogram and 2D echocardiogram results. She was asymptomatic until she presented with features of gallbladder disease and biliary obstruction. Her rare condition was only discovered when further workup was done for an alleged ampullary mass, which was believed to have caused the unsuccessful biliary clearance via ERCP. This suspected neoplasm was shortly proven to be non-existent on imaging and intraoperative visualization, which leads us to conclude that it was indeed the disarray in the location of internal organs that resulted in a failed ERCP. Certainly, situs ambiguous can be deceiving, even in terms of clinical presentations of diseases. Gallbladder and gallstone disease typically presents with right upper quadrant pain;[12] however, this patient complained of epigastric pain due to the aberrant location of her gallbladder at the midline. Hence in such cases, a good clinical eye, together with imaging, becomes highly indispensable.

Laparoscopic approach has been the standard of care in managing gallbladder and biliary diseases. There have already been published case reports highlighting successful laparoscopic cholecystectomies in patients with situs ambiguous.[13] However, given the atypical locations of thoracoabdominal organs in this rare disease, careful

surgical planning is warranted. In this case, the surgical team opted for an open approach due to the anticipated difficulty of a laparoscopic procedure. The anatomic distortion precludes adequate exposure, which poses a high risk for conversion to open surgery.

Conclusion

The unusual location of organs in situs ambiguous misleads clinicians and hinders them from making an accurate assessment and diagnosis. This makes imaging superior over other diagnostic tests in terms of arriving at a diagnosis. Interventions, such as an ERCP in this case, may even have unfavorable outcomes. Choosing the appropriate surgical approach, if indicated, is crucial given the complexities in anatomy. There is a high likelihood that the surgical team will encounter technical difficulties intraoperatively and may even push conversion of a laparoscopic procedure to an open approach. While there has already been published testament of successful and safe laparoscopic cholecystectomy for calculous cholecystitis in a patient with situs ambiguous, it is still important to consider and plan for possible intraoperative complications, and the safety of the patient.

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