



Polysplenia Syndrome with Choledochal Cyst and Left-Sided Inferior Vena Cava: A Rare Association

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Clinical History

The patient was born full term to a then 21yo G2P1 (1001) mother via spontaneous vaginal delivery, with no noted fetomaternal complications. At one-month-old, the patient had acholic stools and dark urine, as well as generalized jaundice and abdominal enlargement. On the fifth day of admission, the patient died of sepsis.

Imaging Findings

Cranial, chest, and abdominal CT scans were performed using GE Healthcare Discovery ST (316 mA, 100 kV, 80 mL of Ultravist contrast). The cranial CT scan shows unremarkable findings. The chest CT angiogram showed bilobed lungs (**Figure 1A**) with long hyparterial bronchi (**Figure 1B**) and no demonstrable cardiac anomalies (**Figures 2A and 2B**).

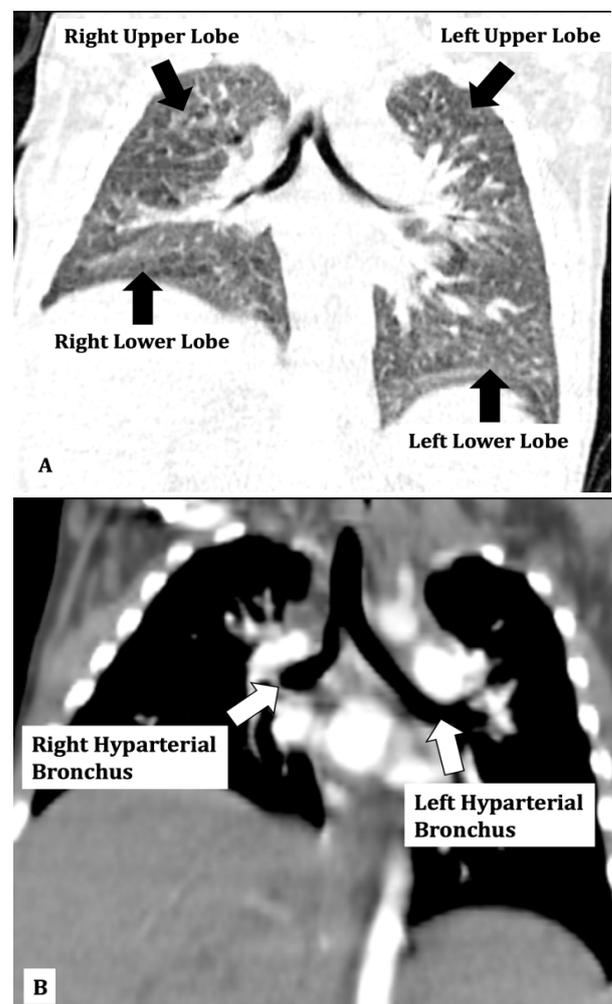


Figure 1: Coronal chest CT sections show bilobed lungs with hyparterial bronchi. Coronal images of the non-contrast CT scan lung window (**A**) and contrast-enhanced (**b**) sagittal exhibiting bilobed lungs, as well as the inferior position of the bilateral bronchi relative to the bilateral pulmonary arteries.



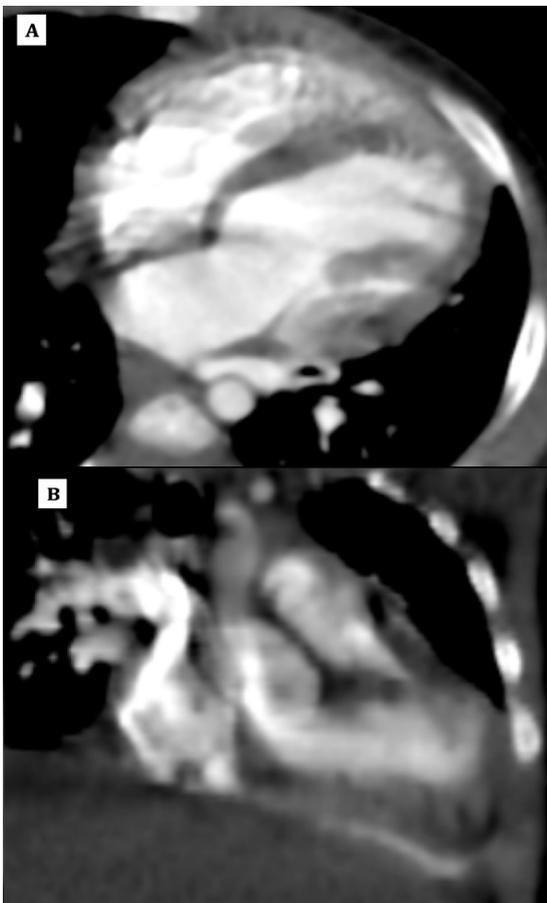


Figure 2: Chest CT shows no cardiac anomalies. Axial (A) and coronal (B) images of a contrast-enhanced CT show no gross evidence of a congenital cardiac anomaly such as septal defects and transposition of great vessels.



Figure 4: 3D reconstruction of the contrast-enhanced CT showing centrally located and enlarged liver.

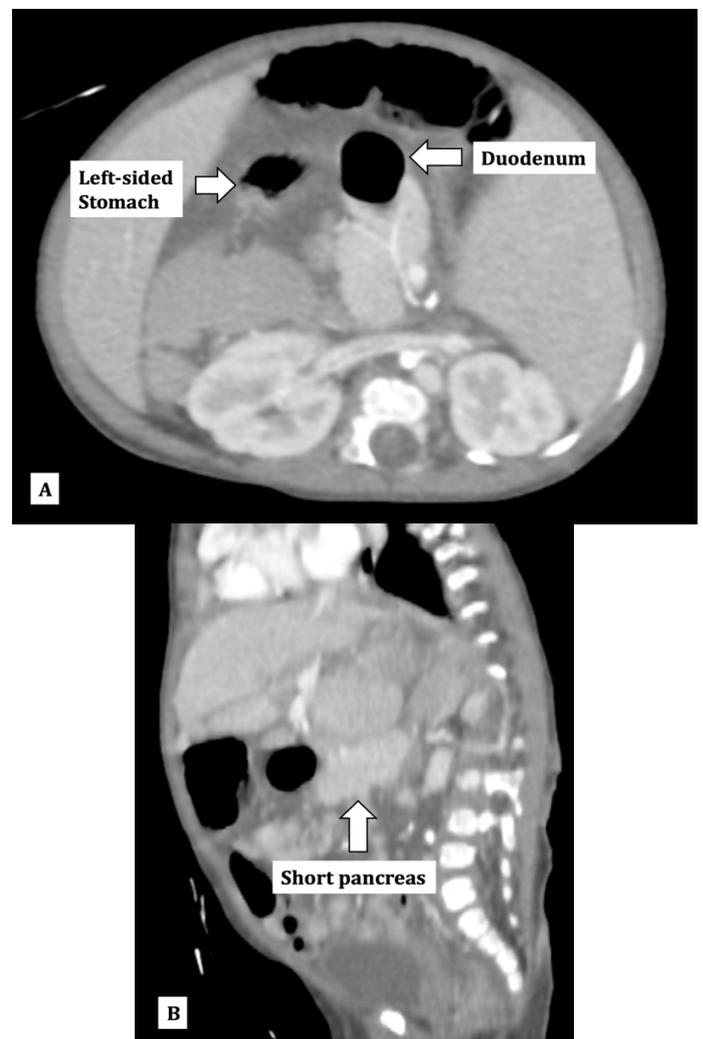


Figure 5: Abdominal CT scan shows pancreatic abnormality. Axial (A) and sagittal (B) images of a contrast-enhanced CT showing a right-sided stomach and short anteroposteriorly positioned pancreas with suspicious incomplete encircling of the proximal duodenum.



Figure 3: Abdominal CT scan shows multiple spleens. Axial (A) and sagittal (B) images of a contrast-enhanced CT showing multiple spleens.

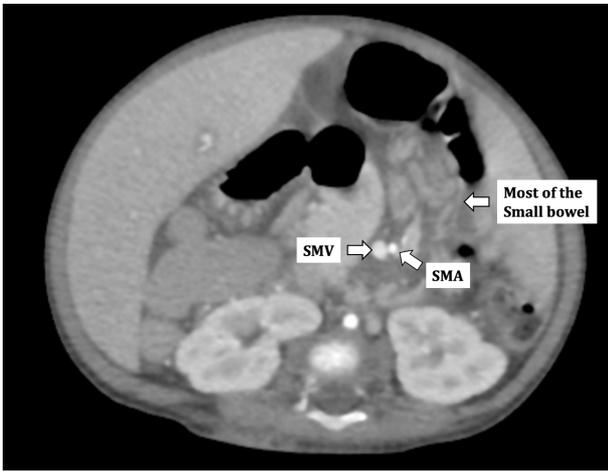


Figure 6: Axial abdominal CT section shows intestinal malrotation. There is an associated abnormal superior mesenteric artery-superior mesenteric vein relationship and non-horizontal duodenum.

The abdominal CT scan shows left isomerism, exhibiting the following findings: right-sided spleen with six surrounding smaller spleens (Figure 3A and 3B), centrally located and enlarged liver (Figure 4), short anteroposteriorly positioned pancreas with suspicious incomplete encircling of the proximal duodenum (Figure 5A-5B), dextro-positioned stomach (Figure 5A), intestinal malrotation with abnormal SMA-SMV relationship and non-horizontal duodenum (Figure 6), left-sided inferior vena cava with primary azygous-hemiazygous drainage (Figure 7A-E), and two right renal arteries (Figure 8). Lastly, there is also focal fusiform dilatation of the common bile duct (Figure 9A), with upstream dilatation of the intrahepatic ducts (Figure 9B).

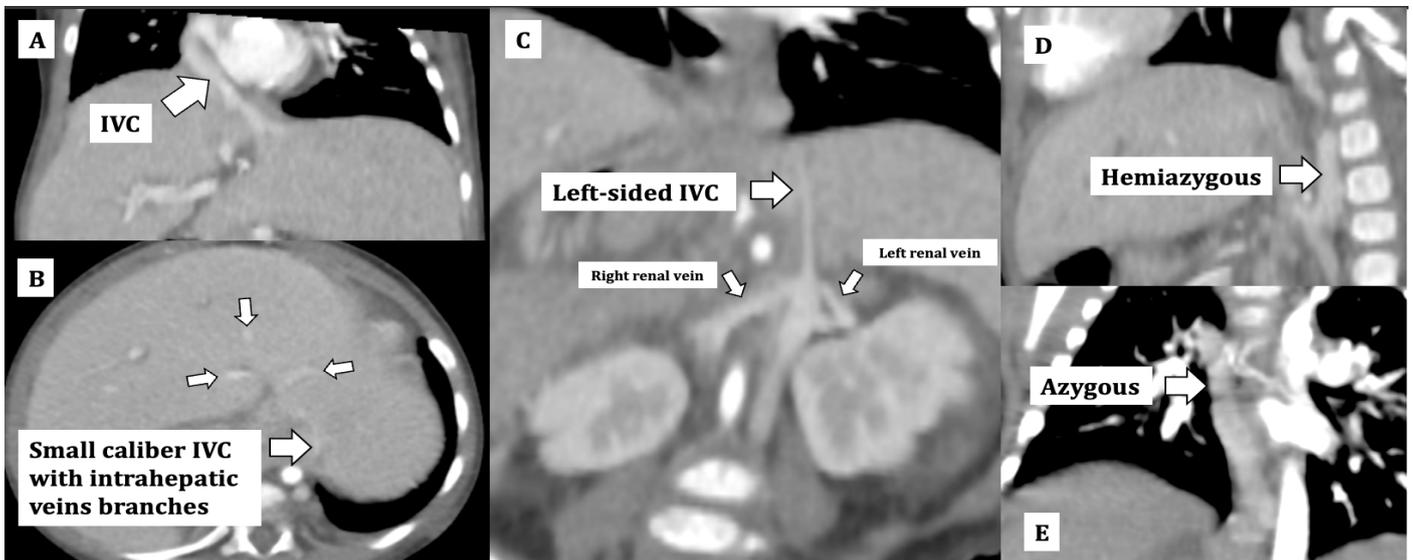


Figure 7: Chest and abdominal CT scan showing a left-sided inferior vena cava. Coronal, axial, and sagittal CT images showed a small caliber left-sided inferior vena cava (A and C) with primary azygous-hemiazygous drainage (D and E). The intrahepatic veins and both renal veins drain into the left-sided IVC.

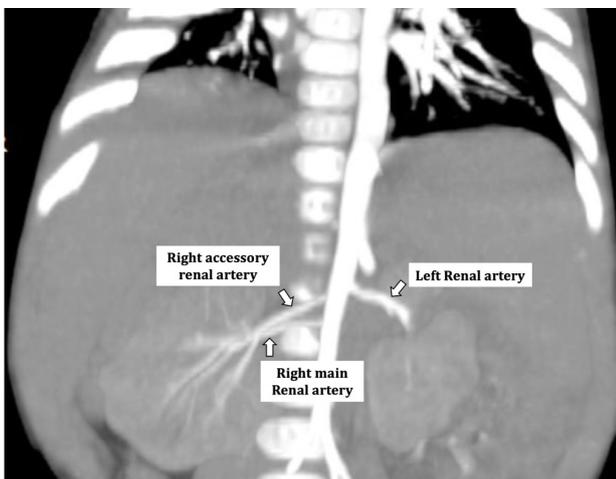


Figure 8: Maximum intensity projection CT image highlighting the two right renal arteries arising from the abdominal aorta.

Discussion

Background

The normal orientation of organs with respect to the midline is described as situs solitus. On the other hand, organs that are in mirror-image orientation are called situs inversus [1]. The case of this patient does not conform to either of these but falls under heterotaxy syndrome or situs ambiguus. Heterotaxy syndrome can be classified into asplenia (right isomerism) and polysplenia (left isomerism) [2]. There is no known single etiologic factor or even chromosomal aberration that can explain the development of abnormal lateralization and isomerism [3]. Recent studies showed that more than 80 genes are required for normal left-right organ development, some of which include ZIC 3, NODAL, LEFTY 2, and SHROOM 3 [4].

Clinical Perspective

The precise incidence of isomerism is not known but most of the early diagnosed cases are right isomerism, due to the early presentation of its associated severe congenital anomalies.

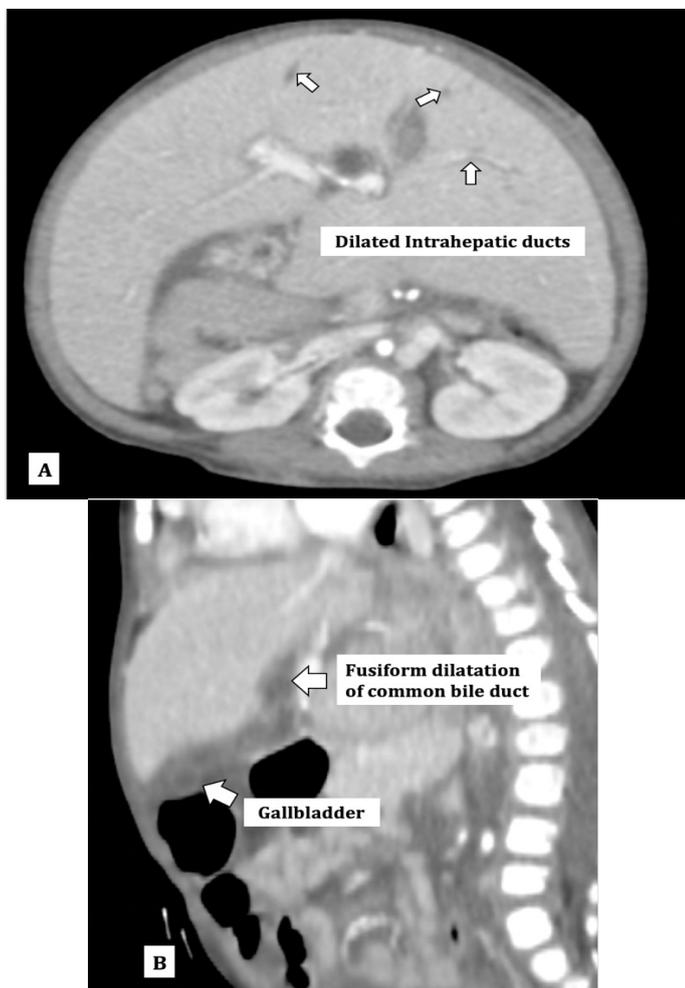


Figure 9: Abdominal CT axial and sagittal images show a focal fusiform dilatation of the common bile duct (B), with upstream dilatation of the intrahepatic ducts (A). The gallbladder is intact and is antero-inferiorly directed (B).

Polysplenia is considered a rare syndrome, with an incidence of 1 per 250,000 live births [5]. About 50-90% of these patients have complex cardiac anomalies and only 10% reach adulthood without any complications [1]. Its clinical presentation is highly dependent on the associated congenital anomaly, which is why adequate imaging is needed to assess these for proper clinical/surgical management. CT and MRI are the best modalities for characterizing the diverse visceral and vascular abnormalities of heterotaxy syndrome [5].

Imaging Perspective

There are no fixed pathognomonic features of polysplenia syndrome [5]. The majority of the findings commonly seen in these patients are bilobed lungs with both hyperarterial bronchi, multiple spleens, intestinal malrotation, short pancreas, midline liver, and anomalies of the inferior vena cava [1]. All of these findings are present in our patient but what is peculiar in this report is the choledochal cyst and a left-sided inferior vena cava with a primary hemiazygous-azygous drainage [5].

Outcome

Management of polysplenia syndrome is mainly symptom-specific, with the majority of treatment being focused on the correction of cardiac anomalies. Severe cardiac anomalies in patients with polysplenia syndrome have a poor prognosis, with 75% of patients dying before age of five [6]. Furthermore, the presence of intestinal malformation and in our case choledochal cyst also warrants surgical intervention as these may cause bowel obstruction and cholangitis, respectively.

Take Home Message/Teaching points

Polysplenia or left isomerism is a rare heterotaxy syndrome associated with multiple congenital anomalies. Its key features are multiple spleens with bilobed lungs and anomalies of other asymmetric organs. Complete imaging work-up of these patients is important since the prognosis of this disease depends on the severity of its other associated anomalies, particularly cardiac, vascular, and gastrointestinal.

Written informed patient consent for publication has been obtained

Final Diagnosis

Polysplenia syndrome with situs ambiguous, left-sided IVC, and choledochal cyst

Differential diagnosis list

- Situs inversus – ruled out since the bronchial anatomy corresponds to the atrial situs and there is a mirror image of the structures on the left side of the chest
- Asplenia syndrome – ruled out due to the presence of spleen
- Kartagener syndrome – ruled out since this is not situs inversus associated with bronchiectasis and sinusitis

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