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An Incidental Finding of Situs Ambiguus with Polysplenia in an Asymptomatic 70-Year-Old Male

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ABSTRACT

The term "Situs ambiguus" describes the configuration when some, but not all thoracic and abdominal organs are abnormally positioned. It is commonly associated with polysplenia, or multiple spleens. 50-100% of situs ambiguus with polysplenia have associated cardiac malformations [1]. We present a 70 year old male who came to our hospital for complete, acute, urinary retention, and was found to have invasive prostate cancer. Incidentally, the liver was positioned on the left side, and the spleen was located on the right side. There were multiple perisplenic nodular formations with peripheral calcifications that had enhancement kinetics similar to those of the spleen, suggesting polysplenia. In low thoracic cuts, the tip of the heart was clearly on the left side. The presence of situs ambiguus with polysplenia in the absence of cardiac abnormalities makes this case a unique presentation of an already rare condition. Clinicians should regularly consider the possibilities of anatomical variation in their medical and surgical practices.

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Introduction

Situs anomalies are important to recognize, as atypical anatomy may pose challenges to clinicians in accurately assessing and treating patients. Situs anomalies are frequently found in children while being worked up for concomitant conditions such as congenital heart disease, immune deficiency, and bowel malrotation with obstruction [1]. On the other hand, situs anomalies in adults are often found as incidental findings, during workups for appendicitis or disease of the gallbladder [1].

It is important to understand specific terminology to describe common anatomic findings. Situs solitus describes the normal configuration of the heart and abdominal organs, and is rarely associated with cardiac abnormalities [2]. To the contrary, situs inversus refers to all visceral organs opposite their expected side relative to midline [2]. This configuration may be described as with dextrocardia or levocardia, with the heart positioned on the right or left, respectively [2]. Further, situs ambiguus, also known as heterotaxy, describes the configuration when some, but not all thoracic and abdominal organs are abnormally positioned [2]. Based on splenic morphology, situs ambiguus may be further classified as either with polysplenia (multiple spleens) or asplenia (absence of a spleen) [2].

The phenotypic diversity of situs ambiguus makes epidemiologic review difficult. Methodological differences in inclusion criteria between published studies pose challenges in estimating prevalence. Additionally, asymptomatic patients may never be diagnosed, potentially yielding underestimates in prevalence data. Data from large studies vary, but tend to estimate prevalence of 1 in 10,000 worldwide [3]. Lopez et al. report 70% higher rates of heterotaxy in infants to Hispanic or black mothers [4].

While the causes of heterotaxy are largely unknown, genetic etiologies and maternal environment are likely to contribute[3]. Growing evidence hints that the spectrum of disorders may have a genetic component, as studies have shown an x-linked inheritance of heterotaxy after manipulation of the zinc finger transcription factor ZIC3 located on chromosome Xq26.3 [5]. Additionally, mothers with diabetes were found to be three times more likely to have a child with heterotaxy as compared to mothers without diabetes [4].

Despite the causes being poorly understood, there are patterns to the anatomical presentations of patients with heterotaxy. These patterns, among many other documented organ malformations, usually result in situs ambiguus being discovered in infancy or childhood, making the following case we present even more intriguing [2].

Case Presentation

We present a 70 year old male with history of DM (on insulin) who presented to our hospital several times for complete, acute, urinary retention, preceded by dysuria, weak urinary stream, and incomplete voiding of the bladder. On first admission to our service, digital rectal exam revealed an enlarged prostate with no appreciable nodularity. Ultrasound revealed a prostate estimated

to be 81.1cc, with normal findings of the bilateral kidneys and the bladder. Urine culture was positive for non-fermenting bacilli. A CT scan with contrast revealed an enlarged, heterogenous enhanced prostate, with evidence of invasion of the posterior bladder wall. The prostate also encroached the anterior wall of the rectum with no obvious signs of invasion. There was evidence of right lateral peri-lesional lymphadenopathy.

Incidentally, the liver was positioned on the left side. The spleen was located on the right side, with calcifications at the periphery. There were multiple perisplenic nodular formations with peripheral calcifications that had enhancement kinetics similar to those of the spleen, suggesting polysplenia. In low thoracic cuts, the tip of the heart was clearly on the left side. There was no evidence of peritoneal effusion. The pancreas, kidneys, and adrenals appeared normally configured.

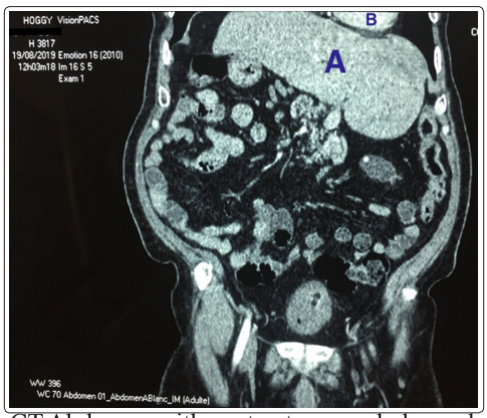
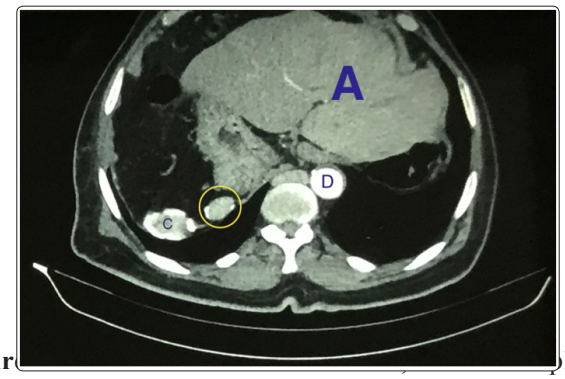


Figure 1: CT Abdomen with contrast, coronal plane, showing the liver (A) and the inferior edge of the heart (B) on the left hand side.



Figur planes, depicting the liver (A) on the left, the spleen (C) on the right with peripheral calcifications, and the aorta (D). There is a perisplenic nodular formation (yellow circle) with peripheral calcifications and enhancement kinetics similar to the spleen, suggestive of polysplenia.

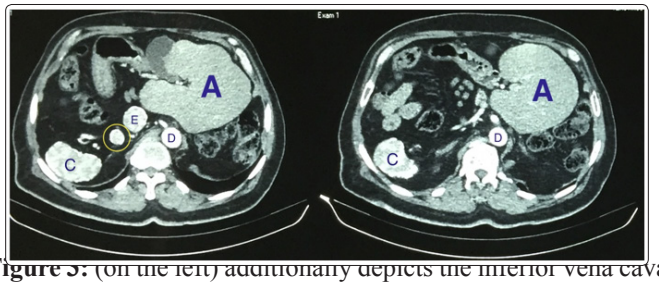


Figure 3: (on the left) additionally depicts the inferior vena cava (E), in addition to the previously described liver (A), spleen (C), perispenic nodules (yellow circle) and aorta (D). Figure 3.2 (on the right) lacks the presumed inferior vena cava.

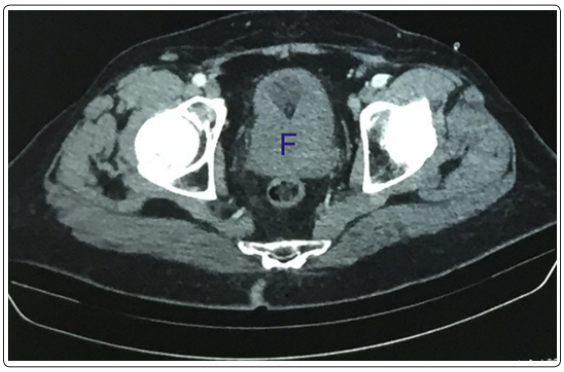


Figure 4: CT Pelvis with contrast, transverse plane, depicting the prostate (F) invading the bladder wall anteriorly. Posteriorly, the border of the prostate approaches the rectum without obvious signs of invasion

Outcome and Follow Up

The patient's abnormal anatomy did not complicate or alter his management. His urologic conditions were managed surgically, and his postoperative course was uneventful. The patient is still living.

Discussion

As depicted in figures 2 and 3, and per the radiology report, the spleen is located on the right side with peripheral calcifications (C). There are multiple perisplenic nodular formations with peripheral calcifications (circled in yellow) with enhancement kinetics similar to those of the spleen, suggesting the presence of polysplenia. While biopsy of the tissue would be needed to confirm the presence of splenic tissue, this was not performed due to the asymptomatic nature of this patient with respect to the spleen. These abdominal CT findings exclude the possibility of asplenia, as is often found with situs anomalies. The patient additionally had no history of recurrent infections.

Congenital heart disease occurs in less than 1% of patients with situs solitus, 3-5% of patients with situs inversus, and has been estimated to be present in 50-100% of individuals with situs ambiguous [1]. Interestingly, the severity of concomitant cardiac malformations seems to vary based on the splenic morphology associated with situs ambiguus. Situs ambiguus with asplenia tends to be associated with cardiac malformations such as total anomalous venous return, atrioventricular septal defects, and bilateral superior venae cavae [1]. In contrast, situs ambiguus with polysplenia is more commonly associated with vena caval interruption with azygous continuation [1,6]. Although there was no thoracic imaging available for our review, this patient's advanced age, lack of a past cardiac medical history, and the presence of a left sided cardiac apex on low thoracic cuts suggest that this situs ambiguus occurred in the absence of concomitant cardiac malformations, which is a rare presentation.

In examining 19 adults, Fulcher & Turner (2002) found that cardiac abnormalities in polysplenia are less severe than those in asplenia, offering a better long-term prognosis, which correlates with this patient's longevity [1]. Along with cardiac malformations, situs ambiguus typically presents with several other organ anomalies. Examples include bilateral bilobed or trilobed lungs, vertebral anomalies, neural tube defects, and urinary anomalies such as renal hypoplasia, horseshoe kidney, and posterior urethral valves [2]. In this case, the radiology report detailed no evidence of kidney, adrenal, or pancreas abnormalities.

Conclusion

Given the growing evidence that situs ambiguus with polysplenia is commonly associated with several, often life-threatening, malformations, more extensive imaging such as a thoracic CT, echocardiogram, and CT angiogram would have provided further insight into our patient's condition. His lack of related medical history and advanced age, however, point to a unique presentation of an already rare condition. Clinicians should regularly consider the possibilities of anatomical variation in their medical and surgical practices. Documentation of these cases is important for both developing a more comprehensive approach to understanding patient presentation and management of patients with abnormal anatomy.

L**earning Points/Take Home Messages**

- Situs ambiguus describes the configuration when some, but not all thoracic and abdominal organs are abnormally positioned. It is commonly associated with polysplenia.
- As 50-100% of situs ambiguus with polysplenia cases have associated cardiac malformations, this case is unique due to the lack of cardiac abnormalities, perhaps explaining how late in life these findings were discovered.
- Clinicians should regularly consider the possibilities of anatomical variation in their medical and surgical practices.

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