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A case report of situs ambiguous with polysplenia and left-sided isomerism

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Abstract

A 78-year-old woman had a staging computed tomography (CT) after the histological diagnosis of sigmoid colon adenocarcinoma. The CT showed situs ambiguous and right-sided polysplenia. In addition to a left-sided liver, there was inferior vena cava interruption with azygos vein continuation. The lungs were bilobed with an absence of the middle lobe, also known as left isomerism.

Keywords

situs ambiguous; heterotaxy; polysplenia; inferior vena cava interruption

Introduction

Situs ambiguous, also known as heterotaxy syndrome, is the abnormal positioning of thoracic and abdominal organs and vessels to that found in situs solitus [1]. This results from errors in left-right patterning during embryogenesis [2].

The occurrence of situs ambiguous happens in roughly 1 per 40,000 live births with a male:female ratio of 2:1 [3]. It should be recalled that this situs abnormality does not have a fixed set of characteristics that are present in all cases [1]. However, there are two major clinical sub-divisions within situs ambiguous. One group are patients with situs ambiguous and polysplenia and left isomerism and these patients only have two bilateral lung lobes. The other group are patients with situs ambiguous and asplenia who have right isomerism with three bilateral lung lobes [4].

Case Presentation

A 78-year-old female had recently had a left total hip replacement, and was on rivaroxaban as a prophylaxis for venous thromboembolism, when she had an episode of rectal bleeding. The bleeding settled without any intervention and she was referred for an urgent endoscopy. During the endoscopy, a distal sigmoid lesion was biopsied and was diagnosed on histology as a moderately differentiated adenocarcinoma with surface dysplasia.

It was known that she had dextrocardia after a chest radiograph was carried out as part of the pre-operative assessment. As shown in the chest radiograph (figure 1), the stomach bubble was not visible so it was not known at that time whether there were further situs anomalies.

She has type 2 diabetes.

A computed tomography scan of the thorax, abdomen and pelvis was carried out as part of the staging protocol. There was the incidental finding of situs ambiguous with multiple spleens of variable sizes on the right side. The liver was left-sided with the presence of inferior vena cava (IVC) interruption with azygos continuation. The hepatic veins drained directly into the left atrium. The azygos vein was dilated and drained into the left atrium via the superior vena cava.

The colon was totally right-sided with the small bowel present entirely on the left side. In addition to the dextrocardia (figure 2), there were two lung lobes bilaterally with the absence of a middle lobe.

She was admitted for a high anterior resection. She developed persistent nausea and vomiting post-operatively and was diagnosed with paralytic ileus. She made a good recovery.

Discussion

Situs anomalies have been described mainly in the paediatric population because these anomalies are strongly associated with congenital heart disease, immunodeficiency and intestinal obstruction which prevent many of the patients reaching adulthood [4]. This has resulted in a lack of literature about these entities in adults.

As described, case reports of situs ambiguous in elderly patients are rare. The patient described in the case report has situs ambiguous with polysplenia and left isomerism (figure 3). She had no history of congenital heart disease, which explains her survival into adulthood.

In situs ambiguous, 90-99% of the patients have severe congenital heart disease. In patients with asplenia, this results in a mortality rate of 79% in the first year and for patients with polysplenia, the mortality rate is 61% in the first year [2]. Only 5% of patients with polysplenia survive into adulthood.

The aetiology of situs ambiguous is poorly understood however certain genes and neurotransmitters have been identified as being vital in the development of left-right axis in the fetus. Mutations in the zinc finger transcription factor (ZIC3) is a cause of X-linked heterotaxy [5]. Since this gene is located on the X chromosome, it is only seen in male offspring. ZIC3 mutations have been associated with familial cases, however it is still unclear whether ZIC3 mutations also contribute to sporadic cases of situs ambiguous [3].

Furthermore, the neurotransmitter serotonin (5-HT) is vital in establishing laterality in the fetus. Interruption of 5-HT signalling is known to result in cases of situs inversus and ambiguous, dextrocardia and a huge range of congenital heart conditions [5].

Interrupted IVC has been reported to be present in 80% of patients with situs ambiguous and polysplenia [6]. This means that the IVC was replaced by the azygos vein on the left and an accessory hemiazygos vein on the right side in the retrocrural space and both showed a certain degree of dilation (figures 4 and 5). A large venous trunk forms in the liver from the hepatic veins and this drains directly into the atrium. In our patient, this was the left atrium due to the presence of dextrocardia.

This case shows that situs ambiguous is not always associated with morbidity and can present in adults as an incidental finding. It reinforces the concept that situs abnormality does not always cause symptoms or medical problems in adults. These anomalies can result in confusion during invasive procedures and it is necessary to perform careful analysis of these anomalies prior to any surgical or

medical interventions.

Through the use of radiology, these situs anomalies are being detected more frequently and most likely will be detected with higher frequency in the future [7].

Figures

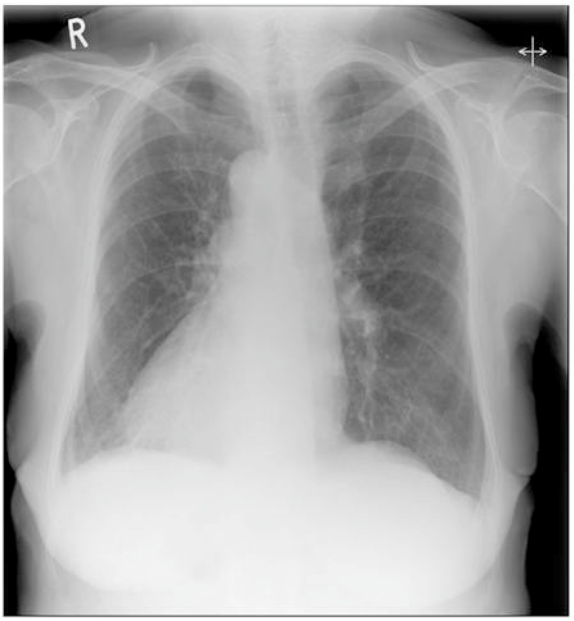

Figure 1: Dextrocardia is demonstrated on this frontal chest radiograph.

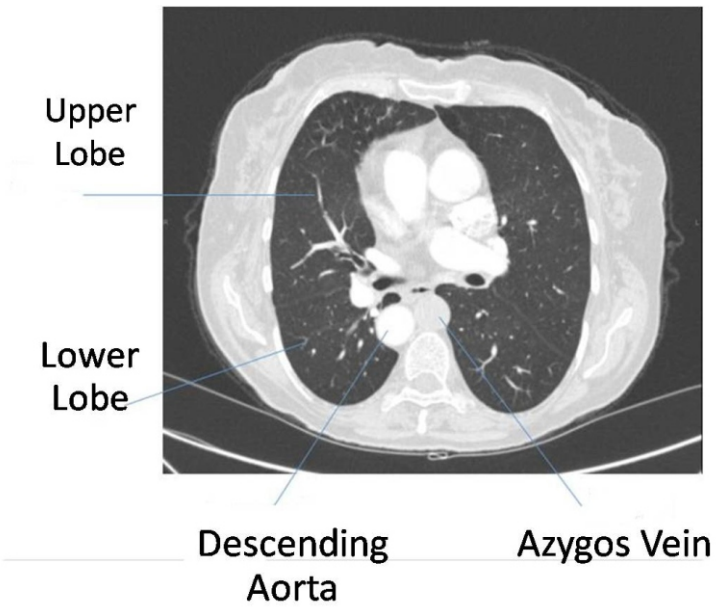

Figure 2: Axial single-slice computed tomography through the chest. Dextrocardia is again demonstrated. The azygos vein is seen on the left side and is prominent. The lungs are bilobed bilaterally and the middle lobe is absent.

Figure 3: Coronal single slice computed tomography through the upper abdomen. The liver is present on the left side and there are three spleens on the right side. The colon is located totally on the right side.

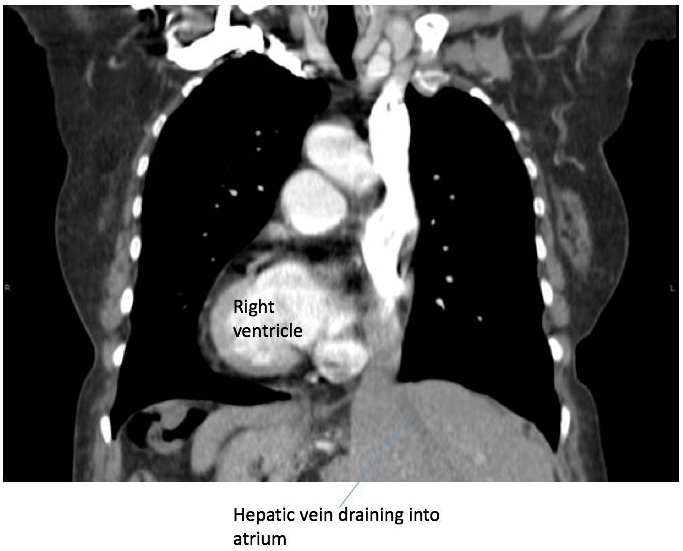

Figure 4: The hepatic veins drain directly into the left atrium. The right ventricle has a thicker muscular wall compared to the left ventricle.

Figure 5: The hepatic segment of the IVC is absent so the hepatic veins join and drain directly into the left atrium. The IVC continues as a dilated left-sided azygos vein.

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