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Heterotaxy syndrome: A rare case report

Imrani Kaoutar, Ola Messaoud, Hounayda Jerguigue, Rachida Latib, Youssef Omor

ABSTRACT

Heterotaxy syndrome is a spectrum of abnormalities that should be differentiated from situs inversus. Multiple spleens are the most common malformation seen in this syndrome. Intestinal rotation abnormalities should always be looked for, because of the risk of midgut volvulus that can be prevented by a correction procedure. We report the case of a 43-year-old patient with heterotaxy syndrome.

Keywords: Heterotaxy, Imaging, Malformation, Polysplenia

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INTRODUCTION

Heterotaxy syndrome or situs ambiguus is a rare disease that should be differentiated from situs inversus. It is characterized by a spectrum of abnormalities that associates with malformations of multiple organs and vessels, with or without anomalies in their arrangement across the left and right axis of the body [1–3]. The diagnosis is made by the association of a sufficient number of malformations that can be easily demonstrated by radiology imaging, such as ultrasound, computed tomography (CT) scan, or magnetic resonance imaging (MRI) [3, 4]. Discovered at young age when critical anatomic malformations are associated, such as biliary atresia or cardiac defect. The knowledge about these malformations is crucially important to set the diagnosis, to look for complication and prevent them if predisposed factors existed.

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CASE REPORT

A 43-year-old woman with no relevant clinical history presented to the hospital complaining of atypical abdominal pain without fever or other associated signs. The physical examination revealed a mass in the left hypochondrium. Ultrasound was realized which showed liver in the left hypochondrium with a pseudotumoral hydatid cyst. Enhanced abdomino-pelvic CT scan at the portal phase was performed to search for other abdominal anomalies, revealing abdominal situs ambiguus with a liver at the left hypochondrium, polysplenia at right hypochondrium, short pancreas (Figure 1), complete

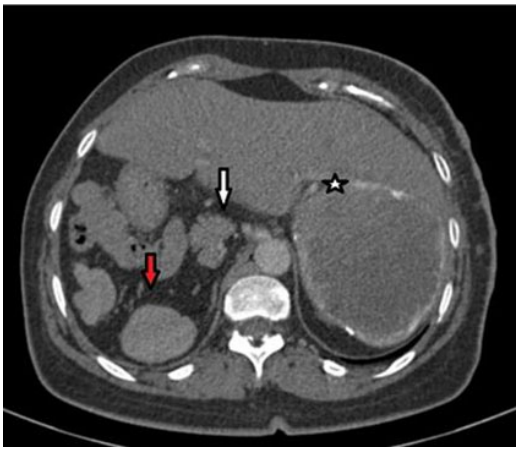

Figure 1: Enhanced abdomino-pelvic CT scan at the portal phase showing abdominal situs ambiguus with short pancreas (white arrow) and polysplenia (red arrow). Note liver hydatid cyst (star) which was incidentally discovered in our patient.

common mesentery (Figure 2). The inferior vena cava was agenesis above the renal veins (Figure 3) with a retroaortic left renal vein (Figure 4). The azygos vein was enlarged substituting for the inferior vena cava (Figure 5). Hepatic veins were flowing directly into the right atrium (Figure 6) and the superior mesenteric artery was localized behind the superior mesenteric vein (Figure 7). Preduodenal portal vein and cardiac anomalies were not found in this case.

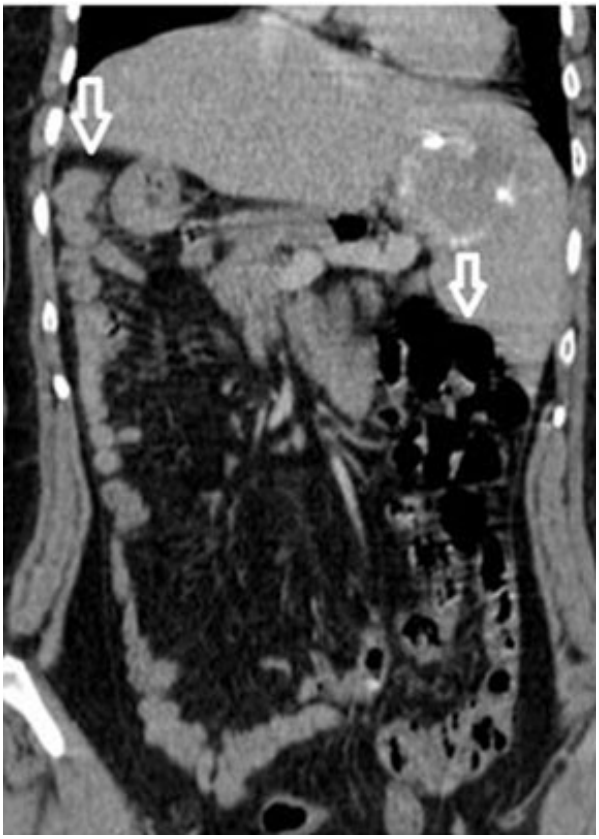

Figure 2: Enhanced abdomino-pelvic CT scan at the portal phase showing complete common mesentery (arrow).

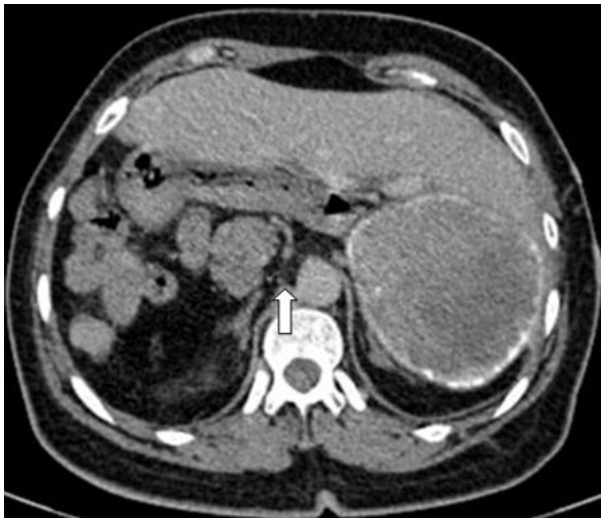

Figure 3: Enhanced abdomino-pelvic CT scan at the portal phase showing inferior agenesis vena cava above the renal veins (white arrow).

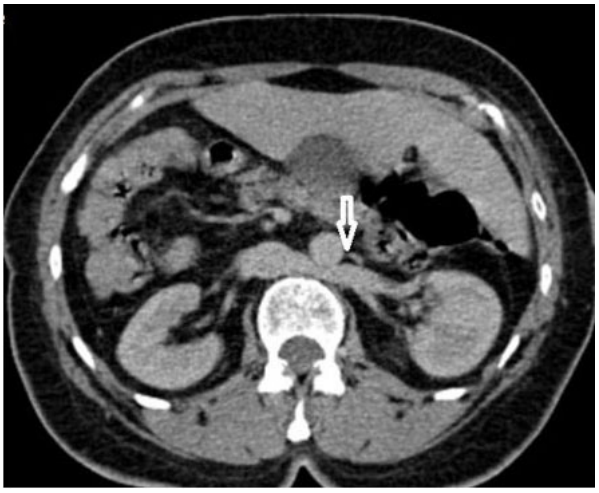

Figure 4: Enhanced abdomino-pelvic CT scan at the portal phase showing a retroaortic left renal vein (arrow).

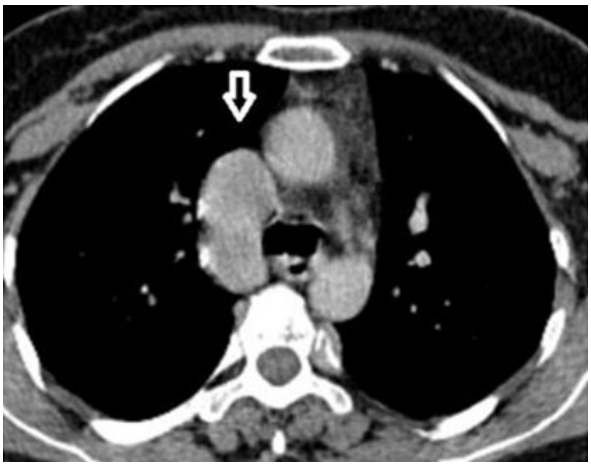

Figure 5: Enhanced abdomino-pelvic CT scan at the portal phase showing enlarged azygos vein substituting for the inferior vena cava (arrow).

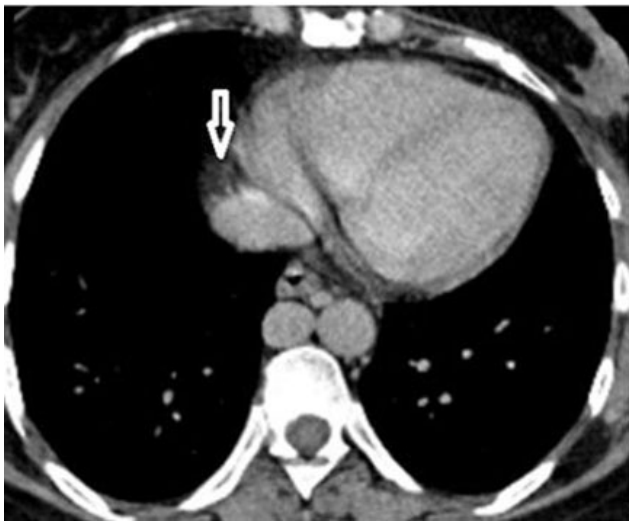

Figure 6: Enhanced abdomino-pelvic CT scan at the portal phase showing hepatic veins flowing directly into the right atrium (arrow).

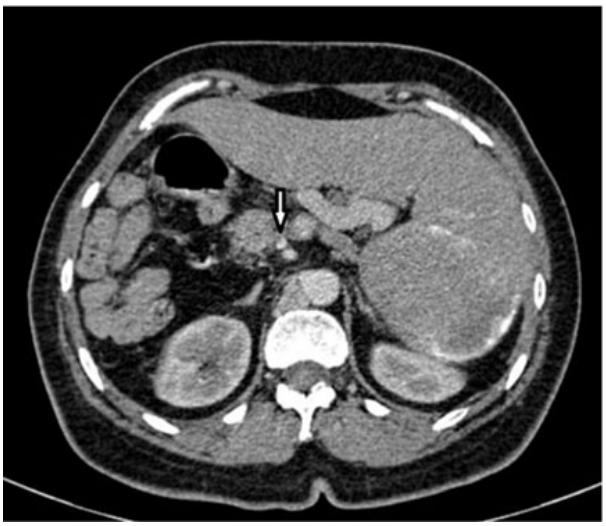

Figure 7: Enhanced abdomino-pelvic CT scan at the portal phase showing the superior mesenteric artery behind the superior mesenteric vein (arrow).

DISCUSSION

Heterotaxy syndrome is a rare disease characterized by a spectrum of abnormalities that we can categorize into two subtypes asplenia and polysplenia syndrome, unlike situs inversus which is a one set of abnormalities.

Situs inversus is a genetic condition in which the organs in the chest and abdomen are positioned in a mirror image from their normal positions.

It is called situs inversus totalis when there is a total transposition of abdominal and thoracic viscera.

Heterotaxy syndrome associates with malformations of multiple organs and vessels, associated or not with anomalies in their arrangement across the left and right axis of the body [1, 3]. Usually discovered in an early age because of critical anatomic malformations such as biliary atresia or a cardiac defect. It can be discovered incidentally in adulthood in patients with mild anatomical malformations, as in the present case [4].

The 1-year mortality in patients with heterotaxy syndrome with complex cardiac lesion is 85% for patients with asplenia and 50% for patients with polysplenia [2].

The diagnosis of polysplenia syndrome is made by the association of a sufficient number of malformations that can be easily demonstrated by radiology imaging, such as ultrasound, CT scan, or MRI [3, 4].

Multiple spleens are the most frequent abnormality; however, some cases have been described with a single bilobed spleen or a single normal splenic gland. The spleens can be right or left sided but, in all cases, are located on the same side of the stomach along the great curvature. Different diagnoses should be known, such as accessory spleen or splenunculus, small nodules of spleen usually <1 cm separated from the rest of the organ, and abdominal splenosis in a post-traumatic or surgical context that refers to an auto-implantation of small deposits of spleen in the abdomen [3].

The second most common abnormality is the interruption of the inferior vena cava with azygos or hemi azygos continuation, it was found in seven of eight patients in the study of Fulcher et al., whereas the suprahepatic veins drain in the atrial cavity. The infrahepatic inferior vena cava can be either right-sided, left-sided, or duplicated [3, 4].

Anomalies of the abdominal aorta location can be associated, in some cases, with infrahepatic vena cava duplication, the aorta is located on the right side of midline between the right and the left infrahepatic vena cava [3]. In the present case the abdominal aorta was normally located on the left side of the midline.

Intestinal rotation abnormalities can be associated to polysplenia, more frequently with patients who suffer from abdominal heterotaxy. These malformations include nonrotation, where the colon is located on the left of the spine and the small bowl on the right, incomplete rotation, with an imaging finding between normal and nonrotation, and the reversed complete and incomplete rotation that is rare [3]. These malformations predispose to midgut volvulus and justify a surgical correction regardless of age and symptoms [5]. Hence, it is necessary to identify this particular abnormality, even if the patient is asymptomatic.

Preduodenal portal vein can be related to this syndrome. It appears as an L-shaped portal vein that is usually asymptomatic but important to notify before biliary or hepatic surgical procedures [6].

Pancreas anomalies have been usually described in heterotaxy syndrome. A high incidence of short pancreas or truncated pancreas was reported, illustrated by the presence of the pancreatic head with or without the pancreatic body. The opacification of a short portion of the pancreatic duct at endoscopic retrograde cholangiopancreatography associated with other malformations that are seen in heterotaxy is referred usually to a truncated pancreas [3]. This malformation increases the risk of pancreatitis and diabetes mellitus [1].

Cardiac anomalies are mostly discovered at a young age. They include atrial septal defect, ventricular septal defect, bilateral superior vena cava, right-sided aortic arch, partial anomalous pulmonary venous return, transposition of the great arteries, pulmonary valvular stenosis, and subaortic stenosis [7]. None of these malformations were found in the present case, which explains the survival of the patient into adulthood [3].

The treatment depends on the specific organs that are affected. Heart surgery may be necessary to correct any heart defects. For some individuals, this may require multiple procedures to correct the defect [1, 6].

CONCLUSION

In the summary, heterotaxy syndrome or situs ambiguus is a rare disease with a spectrum of abnormalities that can be characterized into two subtypes

asplenia and polysplenia syndrome. The knowledge of the abnormalities that can be encountered is fundamental to set the diagnosis and prevent the complications, therefore, it is necessary for the radiologist to be aware of the points given below.

LEARNING POINTS:

- Heterotaxy syndrome is a spectrum of abnormalities that should be differentiated from situs inversus.
- It can be characterized into two subtypes asplenia and polysplenia syndrome.
- Multiple spleens are the most common findings.
- It is necessary for the radiologist to be familiar with this syndrome and have a sufficient knowledge of its malformations and their possible surgical impact, predisposition to certain pathologies and complications.

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Author Contributions

Imrani Kaoutar – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Ola Messaoud – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Hounayda Jerguigue – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Rachida Latib – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Youssef Omor – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Guarantor of Submission

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Consent Statement

Written informed consent was obtained from the patient for publication of this article.

Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.