Heterotaxy syndrome with polysplenia: case report

Síndrome heterotáxica com poliesplenia: relato de caso

Síndrome de heterotaxia con poliesplenia: reporte de caso

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ABSTRACT
Heterotaxy syndrome (HS) or situs ambiguus is a rare condition, more common in women, in which internal organs are abnormally arranged within the thoracic and abdominal cavities. The condition may manifest as asplenia or polysplenia, characterized by the presence of multiple small and dysfunctional spleens. The severity of HS varies depending on the specific abnormalities present. With the increasing use of imaging techniques, incidental findings of polysplenia are likely to become more frequent. The objective of this report is to illustrate typical imaging findings of HS, aiming to familiarize radiologists and physicians with the spectrum of potentially associated anomalies, thereby improving the quality of care for affected patients. This report describes the case of a 53-year-old woman incidentally diagnosed with HS with polysplenia based on chest and abdominal computed tomography (CT).

Keywords: heterotaxy syndrome, congenital abnormalities, splenic diseases.
RESUMO
A Síndrome de Heterotaxia (SH) ou situs ambiguus é uma condição rara, mais comum em mulheres, na qual os órgãos internos estão anormalmente dispostos nas cavidades torácica e abdominal. A condição pode se apresentar como asplenia ou poliesplenia, com a presença de múltiplos baços pequenos e disfuncionais. A gravidade da SH varia de acordo com as anormalidades específicas presentes. Diante do aumento da realização de exames de imagem, é provável que achados incidentais de poliesplenia tornem-se mais frequentes. O objetivo deste relato é ilustrar os achados de imagem típicos da SH, contribuindo para que os radiologistas e médicos em geral estejam familiarizados com o espectro de anomalias potencialmente associadas, objetivando melhoria na qualidade da assistência aos pacientes acometidos. Este relato descreve o caso de uma mulher de 53 anos com diagnóstico incidental de SH com poliesplenia a partir de tomografia computadorizada (TC) de tórax e abdome.

Palavras-chave: síndrome de heterotaxia, anormalidades congênitas, esplenopatias.

1 INTRODUCTION

Heterotaxy Syndrome (HS) or situs ambiguus is a rare condition, more common in females, where internal organs are abnormally arranged in the thoracic and abdominal cavities (1,2). In the abdomen, the condition can present as asplenia or polysplenia, with the presence of multiple small and dysfunctional spleens. The severity of HS varies according to the specific abnormalities present (1).

In most cases, polysplenia is asymptomatic, constituting an incidental finding on imaging exams (3). However, this condition, by altering the arrangement of organs, can lead to confusion during physical examination or when interpreting other medical images, potentially resulting in erroneous diagnoses.

The aim of this HS case report is to illustrate the typical imaging findings of the
condition to raise awareness among the medical community about these anomalies, helping to avoid misinterpretations and ensuring that healthcare professionals recognize these signs when encountered in future cases.

2 CASE REPORT

A 53-year-old woman, diagnosed with human immunodeficiency virus (HIV) infection five years ago and irregularly using antiretroviral therapy due to crack addiction, presented with a two-month history of dry cough associated with approximately 10 kg weight loss during that period, as well as progressive worsening dyspnea, generalized weakness, and chills. She was transferred to the hospital while on antibiotic therapy due to a suspected diagnosis of community-acquired pneumonia or pneumocystosis. Computed tomography (CT) scans of the chest and abdomen were requested due to the possibility of inflammatory infectious alterations, revealing the incidental finding of HS.

3 DISCUSSION

Polysplenia or left isomerism is one of the syndromes of situs ambiguus or heterotaxy. More common in females, polysplenia has a reported incidence of 1 per 250,000 live births (3), and its etiology is still poorly understood. Recent studies in humans have identified genetic mutations in patients with HS, including CFC1 and SHROOM3 (2).

In most cases, polysplenia is asymptomatic, being diagnosed incidentally on imaging exams or in the context of symptoms associated with other concomitant pathologies (3). Although chest radiography and abdominal ultrasonography can facilitate the diagnosis, CT of the chest and abdomen is the best modality for identifying relevant anatomical details in this context (2).

Polysplenia is characterized by the presence of multiple spleens (Fig 1), with numbers ranging from two to sixteen, commonly of similar sizes. In some cases, there may be one or two larger spleens associated with multiple smaller spleens (2). On abdominal CT examination, the patient in question presented polysplenia with left isomerism, characterized by the presence of nodular areas of splenic tissue located in the right hypochondrium.

Polysplenia can also be associated with pancreatic anomalies. Case series reveal a high incidence of short or truncated pancreas, where only the pancreatic head may be identified or it may be associated with a small portion of the pancreatic body (2). These findings are consistent
with the present report, with CT showing a pancreas with a usual-looking cephalic region and an uncharacterized tail, suggesting agenesis (Fig 2). The identification of pancreatic alterations gains clinical relevance due to the higher incidence of pancreatitis and diabetes mellitus (2).

Intestinal malrotation is also described as a finding of HS (3), and was evidenced in the described case, with abdominal CT revealing the duodenum located to the right, without crossing the midline. The cecum was located superiorly in the right hypochondrium, and the transverse colon inferiorly in the pelvis, with partial rotation of the colonic loop to the left, without determining intestinal obstruction at the time of the examination (Fig 3).

In the thoracic cavity, polysplenia may present with bilateral bilobed lungs, bilateral hyperarterial bronchi, and bilateral pulmonary atria (2), as well as cardiac anomalies, which were absent in the present case. Thoracic CT showed signs of inflammatory/infectious processes, the reason for hospitalization (Fig 4). On the other hand, the inferior vena cava continuing with the azygos/hemiazygos, draining into the superior vena cava, was evidenced, a finding reported in a previous review study (3) (Fig 5,6).

Genitourinary malformations, such as renal agenesis, hypoplastic kidneys, and duplication of collecting systems (4), also possible in polysplenia, were not identified.

In this case, the clinical manifestations presented by the patient at the time were not associated with polysplenia. However, the identification of anatomical alterations through imaging exams can contribute to follow-up, assisting in future differential diagnoses and potential invasive procedures (5,6).
Figure 1 - Axial CT scan with contrast showing the liver encompassing the entire diaphragm (F), multiple spleens (arrows) behind the right hypochondrium along with the stomach (E) also on the right side.

Source: Personal Archive

Figure 2 - Axial CT scan with contrast highlighting the cephalic region of the pancreas (arrow) with a usual appearance, while the tail was not characterized, suggesting agenesis.

Source: Personal Archive
Figure 3 - Scout image of the tomography demonstrating colic loops distributed predominantly to the left and small bowel loops in the central and right regions, indicating intestinal malrotation. No points of caliber alteration of the loops were identified, ruling out the possibility of occlusion/subocclusion.

Source: Personal Archive

Figure 4 - Axial (A) and coronal (B) CT scans of the chest demonstrating consolidative process related to inflammatory/infectious process.

Source: Personal Archive
Figure 5 - Coronal Maximum Intensity Projection (MIP) reconstruction (A) demonstrating the inferior vena cava after the confluence of the hepatic veins, with no evident continuation of the inferior vena cava in the abdomen (arrows). Adjacent (B) image of a patient with normal anatomy showing the inferior vena cava (arrowheads).

Figure 6 - Coronal Maximum Intensity Projection (MIP) reconstruction showing enlargement of the azygos vein (arrows), collateral circulation due to vena cava anomaly.
4 CONCLUSION

HS is a complex syndrome that can present with a wide range of abnormalities, including polysplenia. With the increasing use of imaging exams, it is likely that incidental findings of polysplenia will become more common. In this context, it is important for radiologists and physicians in general to be familiar with the spectrum of potentially associated anomalies, aiming to improve the quality of care for affected patients.
REFERENCES


