Case 15673

Heterotaxy syndrome with polysplenia
Published on 21.04.2018

DOI: 10.1594/EURORAD/CASE.15673
ISSN: 1563-4086
Section: Abdominal imaging
Area of Interest: Abdomen Head and neck
Procedure: Diagnostic procedure
Imaging Technique: CT
Special Focus: Congenital Case Type: Clinical Cases
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Patient: 52 years, female

Clinical History:

A 52-year-old woman with history of fibromyalgia, atopic eczema, allergic rhinitis, asthma, hypoacusia, dysphonia, and peripheral venous insufficiency presented to our institution with recent onset of pruritus and arthralgia. Her daughter was diagnosed with systemic lupus erythematosus. Physical examination and laboratory tests excluded an autoimmune disease.

Imaging Findings:

For better clarification of the clinical scenario a thoraco-abdomino-pelvic computed tomography (CT) was requested. CT revealed polysplenia (Fig. 1a and 1b) and several vascular abnormalities (Fig. 1a to 1g). Vascular anomalies included interrupted inferior vena cava (IVC) with azygos and hemiazygos continuation, superior mesenteric vein rotation sign, anomalous origin of hepatic artery from superior mesenteric artery, preduodenal portal vein, precaval right renal artery, left renal vein bifurcation draining to IVC and hemiazygos vein and aberrant origin of the left vertebral artery from the aortic arch. Cardiac and lung anatomy was normal.

Discussion:

Situs anomalies imply a disordered organ arrangement in the chest or abdomen. Situs solitus consists of the usual arrangement of organs and vessels within the body. Situs inversus is an anatomic arrangement that is the mirror image of situs solitus. Situs ambiguous or heterotaxy refers to visceral malposition and dysmorphism associated with indeterminate atrial arrangement [1]. Heterotaxy syndrome can be classified as heterotaxy syndrome with polysplenia (classically with bilobed lungs with hyparterial bronchi, bilateral pulmonary atria, midline liver, multiple spleens and variable location of the stomach) and heterotaxy syndrome with asplenia (classically with trilobed lungs with bilateral minor fissures and eparterial bronchi, bilateral systemic atria, midline liver, absent spleen and variable location of the stomach).

Heterotaxy syndrome with polysplenia has a reported incidence of 1/250000 live births and its cause remains unknown [2]. Only 5-10% of patients without cardiac malformations reach adulthood [2]. Heterotaxy syndrome with polysplenia is a heterogeneous disorder with a wide spectrum of anomalies, such as: cardiovascular anomalies, bilateral bilobed lungs with hyparterial bronchi, interrupted inferior vena cava with azygos (and hemiazygos) continuation, polysplenia, truncated short pancreas, midline liver, preduodenal portal veins, intestinal malrotation, stomach malrotation and biliary atresia [1, 2].

 Interruption of the IVC with azygos and hemiazygos continuation is the most consistent finding in heterotaxy syndrome with polysplenia [1]. Preduodenal portal vein is another frequent anomaly with polysplenia. It is a surgically important anomaly and can produce pressure symptoms on the duodenum and bile duct [2].

In our case some vascular anomalies are rare. Anecdotal reports mention an anomalous origin of hepatic artery
from superior mesenteric artery [2]. Rameshbabu et al reported for the first time a precaval position of single main right renal artery and aortic origin of the left vertebral artery associated with polysplenia [2]. To our knowledge this is the first case of left renal vein bifurcation draining to IVC and hemiazygos vein. Due to the fact that there are almost an infinite number of combinations of possible malformations, all suspected cases should be labelled as heterotaxy syndrome and segmental or individualised approach for describing patients’ specific anomalies should be used [1].

Adult cases of heterotaxy with polysplenia are managed with palliative and symptomatic measures [2].

**Differential Diagnosis List:** Heterotaxy syndrome with polysplenia, Heterotaxy syndrome with asplenia, Situs inversus

**Final Diagnosis:** Heterotaxy syndrome with polysplenia

**References:**


**Figure 1**

**Description:** Axial contrast-enhanced CT shows multiple sphenules (yellow), with proeminent aygos (orange) and hemiazygos veins (red). Absent suprarenal IVC is also noted. **Origin:** Department of Radiology, Hospital Dr. Nélio Mendonça - SESARAM, E.P.E., Madeira, Portugal
Description: Axial contrast-enhanced CT at a lower level displays two spenules (yellow), prominent hemiazygos vein (red), pre-caval right renal artery (blue) and preduodenal portal vein (green). Origin: Department of Radiology, Hospital Dr. Nélio Mendonça - SESARAM, E.P.E., Madeira, Portugal
Description: At a lower level depicts a prominent hemiazygos vein (red) draining part of the left renal vein, an anomalous origin of the common hepatic artery (purple) from the SMA and a preduodenal portal vein (green). Origin: Department of Radiology, Hospital Dr. Nélio Mendonça - SESARAM, E.P.E., Madeira, Portugal
Description: Axial contrast-enhanced CT at a lower level depicts the superior mesenteric vein rotation sign, consisting of the superior mesenteric vein (green) anterior and slightly to the left of the superior mesenteric artery (purple). Origin: Department of Radiology, Hospital Dr. Nélio Mendonça - SESARAM, E.P.E., Madeira, Portugal
Description: Axial contrast-enhanced CT image shows a bifurcated left renal vein draining to the IVC and hemiazygos vein (red arrow). Origin: Department of Radiology, Hospital Dr. Nélio Mendonça - SESARAM, E.P.E., Madeira, Portugal
Description: Axial contrast enhanced CT image shows a bifurcated left renal vein draining to the IVC and hemiazygos vein (red arrow). Origin: Department of Radiology, Hospital Dr. Nélio Mendonça - SESARAM, E.P.E., Madeira, Portugal

Description: Axial contrast-enhanced CT shows aberrant origin of the left vertebral artery from the aortic arch (white arrow). Origin: Department of Radiology, Hospital Dr. Nélio Mendonça - SESARAM, E.P.E., Madeira, Portugal