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Case Report

Heterotaxy polysplenia syndrome in an adult female with complete endocardial cushion defect[☆]

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ABSTRACT

Heterotaxy syndrome is a rare condition characterized by the abnormal arrangement of thoracoabdominal organs across the left-right axis of the body.

It is generally classified as right and left atrial isomerism or asplenia and polysplenia syndrome, even though there are overlaps and uncertainties. The diagnosis of isomerism is typically made by echocardiography. However, multidetector computed tomography and MRI can help in obtaining detailed data on the morphology of the heart, great vessels, the anatomy of the internal organs, and their mutual arrangement that make an accurate diagnosis of heterotaxy syndrome.

The authors present here the imaging findings of the heterotaxy polysplenia syndrome in a 21-year-old female with a complete endocardial cushion defect and a duplicated right renal vein.

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Background

Heterotaxy syndrome (HS) is a group of congenital disorders characterized by the abnormal arrangement of thoracoabdominal organs across the left-right axis of the body that differs from patients with situs solitus and situs inversus [1,2]. It is generally classified as right and left atrial isomerism or asplenia and polysplenia syndrome, even though there are over-

laps and uncertainties [1,3]. Patients with left isomerism, usually have less severe cardiac malformations than those with right isomerism. However, some may have complex cyanotic heart disease, frequently associated with a univentricular atrioventricular connection [4]. The diagnosis of isomerism is typically made by echocardiography both prenatal and postnatal, which provides details of intracardiac anatomy and cardiovascular connections. Other imaging techniques such as ultrasound and CT can be used to obtain better anatomic

Abbreviations: HS, heterotaxy syndrome; CHD, congenital heart disease; MDCT, multidetector computed tomography.

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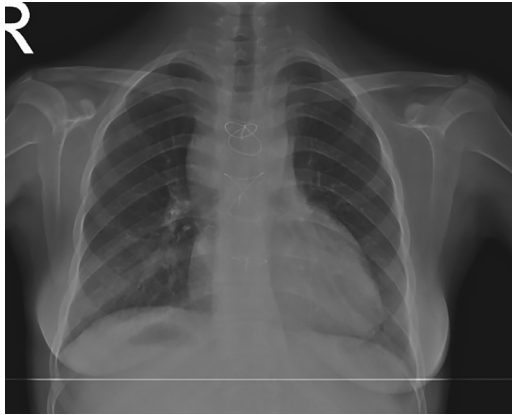


Fig. 1 – The plain chest x-ray image shows wide mediastinum and cardiomegaly.

details and evaluate the location, number, and function of the spleen. MR imaging and cardioangiography are usually performed to establish surgical planning in patients with CHD [5]. The authors present here the imaging findings of the heterotaxy polysplenia syndrome in a 21-year-old female with a complete endocardia cushion defect.

Case presentation

A 21-year-old female was presented to the emergency service of our hospital, complaining of recurrent syncope and head trauma for one day followed by chest pain and numbness of the left arm for one hour. In physical examination, her blood pressure was 90/50 mm Hg, heart rate; 85 beats/min, and she had normal respiratory rate. On auscultation a pansystolic murmur was audible in the precordial region, other findings were unremarkable. She was a known case of endocardial cushion defect, having it since birth. No familial, social, and allergic history was given by the patient.

Her routine blood exams were within normal limits and she was negative for COVID-19. On the plain chest X-ray image, cardiomegaly was observed (Fig. 1). Her brain CT exam was normal and there was no finding for intracranial hematoma or bone fracture. After multidisciplinary consultation, she was discharged from the hospital on necessary medications and close follow-up recommendation.

About one month later the patient was re-presented to the hospital, complaining of fatigue and weakness, and hospitalized for having lower extremities deep vein thrombosis (DVT). The Doppler ultrasound revealed bilateral lower extremities DVT.

On thoracoabdominal CT angiography images, the heart apex was seen on the left. A large defect along the interventricular and interatrial septum was observed, consistent with a complete endocardia cushion defect. The aorta was originated from the ventricle located on the right and the aortic arch was seen on the left (Fig. 2a-b). Both atria were demonstrated in the left atrial morphology with a large interatrial

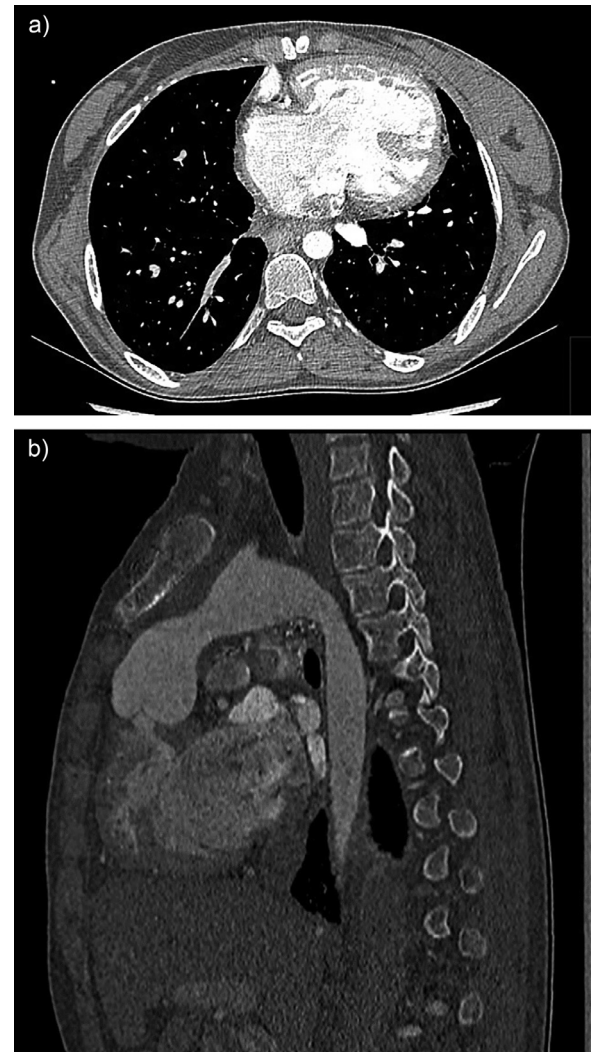


Fig. 2 – (a) Contrast-enhanced axial chest CT angiogram shows both atria in the left atrial morphology, complete endocardial cushion defect, and cardiomegaly. (b) Contrast-enhanced sagittal chest CT angiogram shows that the aorta is originated from the ventricle on the right.

septal defect, resembling the common atrium. The left superior and inferior pulmonary veins were seen to be drained into the atrium located on the right. The inferior vena cava was interrupted and continued with the azygos vein on the left of the abdominal aorta (Fig. 3a-b).

Both lungs were observed as having 2 lobes and bilateral morphological left bronchial anatomy (Fig. 4). The liver filled the left upper quadrant and midline. The right hepatic vein was opened directly into the left atrium. There were multiple spleens (polysplenia) in the right of the abdomen. The cecum was located more medially than normal, compatible with intestinal mal-rotation. The kidney on the right had duplicated veins; the superior vein was draining into the azygos vein and the inferior one drained into the left located inferior vena cava separately, which was unique for this case (Fig. 5a-c).

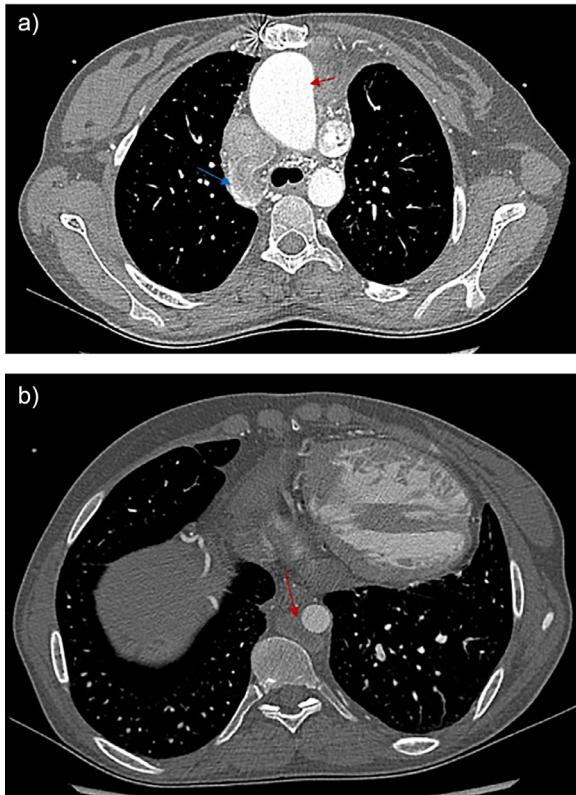


Fig. 3 – (a) Contrast-enhanced axial chest CT angiogram shows the aortic arch on the right (red arrow) and prominent azygos vein continuing on the IVC (green arrow). (b) Interrupted IVC, the continuation of the azygos vein (red arrow).

Discussion

Heterotaxy syndrome or situs ambiguous is defined as an abnormal arrangement of viscera across the left-right axis of the body that occurs in about 0.8% of patients with congenital heart disease [6]. The normal visceral arrangement depends on a series of intricate processes that take place during early mesoderm development, such as adequate expression and leftward flow of growth signals. Impairment in any of these factors during organogenesis may lead to abnormal organ positioning and HS [7]. Recent studies have shown that more than 80 genes are required for normal asymmetric left-right organ development and mutations in some of these genes have been identified in patients with heterotaxy [8].

The HS is generally classified as right and left atrial isomerism or asplenia and polysplenia syndrome, even though there are overlaps and uncertainties [1,3]. HS with asplenia implies that the patient has bilateral trilobed lungs with bilateral minor fissures and eparterial bronchi, bilateral systemic atria, a centrally located liver, and a stomach in an indeterminate position. The abdominal aorta and inferior vena cava would classically be located on the same side of the spine. This syndrome usually occurs in males, and the patients often present with cyanosis and respiratory distress. Whereas, in HS with



Fig. 4 – Coronal chest CT image shows both lungs, having two lobes and bilateral morphological left bronchial anatomy.

polysplenia, the patient has bilateral bilobed lungs, bilateral pulmonary atria, a centrally located liver, a stomach in an indeterminate position, and multiple spleens either in the left or right upper quadrant, along the greater curvature of the stomach. Interruption of the inferior vena cava with azygos continuation may be associated with this type of HS. HS with polysplenia is more common in females and has more variable clinical manifestations and prognosis [9,10]. Patients with left isomerism, in general, have less severe cardiac malformations than those with right isomerism. However, some may have complex cyanotic heart disease, frequently associated with a univentricular atrioventricular connection [4]. Our patient had the characteristic feature of polysplenia or left atrial isomerism, but the complete endocardia cushion defect and duplicated right renal vein was unique for this case.

The diagnosis of isomerism is typically made by echocardiography both prenatal and postnatal, which provides details of intracardiac anatomy and cardiovascular connections. Although the spectrum of anomalies in patients with situs abnormalities is diverse and can be difficult to fully delineate by conventional means. Other imaging techniques such as ultrasound and CT can be used to obtain better anatomic details and evaluate the location, number, and function of the spleen. MR imaging and cardioangiography are usually performed to establish surgical planning in patients with CHD [5,11].

Multidetector computed tomography (MDCT) offers a quicker study and allows to obtain detailed data on the morphology of the heart, great vessels, the anatomy of the internal organs, and makes an accurate diagnosis of HS. However, magnetic resonance imaging offers a radiation-free alternative at the expense of greater study times and the likely need for sedation in infants [12,13]. In our patient, MDCT had an important role in the diagnosis of extracardiac anomalies, the anatomy of the internal organs, and their mutual arrangement.

Treatment of patients with isomerism should be determined by the nature and severity of the associated cardiac and extracardiac lesions. Most cardiac operations for patients

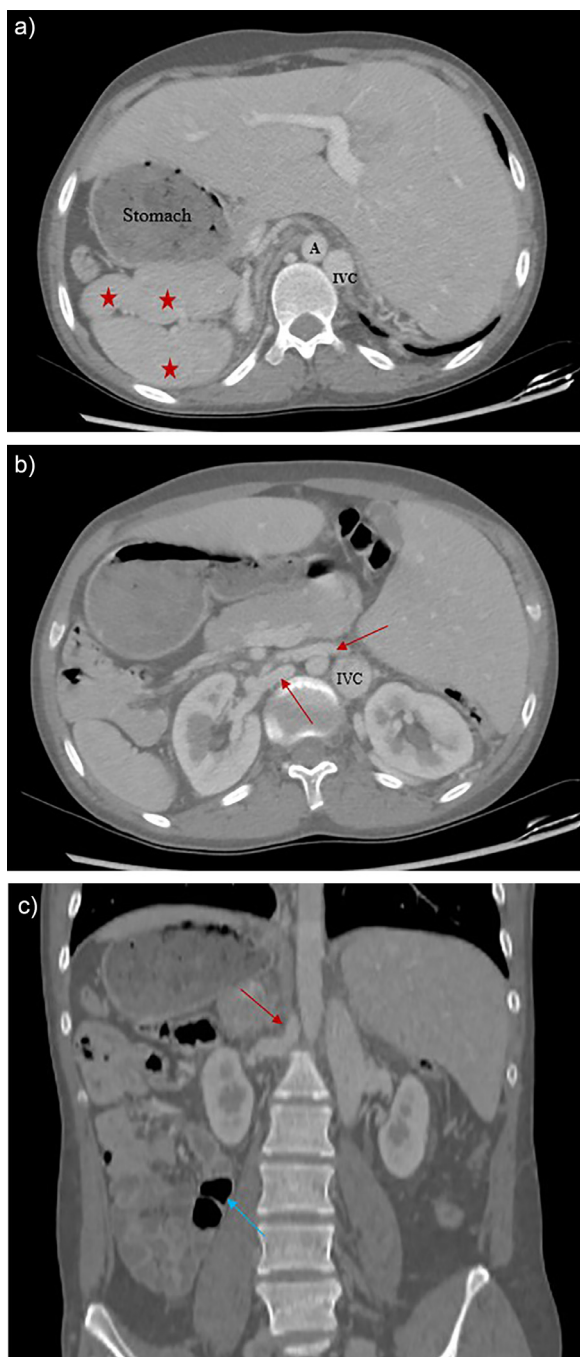


Fig. 5 – (a) Contrast-enhanced axial CT image shows the stomach and multiple spleens (asterisks) on the right and the liver on the left and midline. (b) The right kidney with double veins: the superior vein drains into the azygos and the inferior one drains into the left located IVC (arrows). (c) Contrast-enhanced coronal CT image shows the right kidney with double veins: the superior vein drains into the azygos vein (red arrow), the cecum is located more medially (green arrow), compatible with intestinal malrotation.

with isomerism are palliative in nature since normal anatomy is rarely achieved [4].

Long-term outcomes in HS are poorly described. Some reports suggest improved survival in the recent era, whereas others do not. In general, those with univentricular circulation and totally anomalous pulmonary venous connection have the worst prognosis, while survival is higher in those with biventricular circulation [14]. In our case the patient underwent cardiac surgery 3 times, trying to repair the AV canal defects, though the outcome was unsatisfactory and she is a candidate for heart transplantation.

Conclusion

Heterotaxy syndrome is a rare congenital disorder that is generally classified as right and left atrial isomerism or asplenia and polysplenia syndrome. Patients with left isomerism often have less severe cardiac malformations than those with right isomerism. However, some may have complex cyanotic heart disease, frequently associated with a univentricular atrioventricular connection. The diagnosis of isomerism is typically made by echocardiography, but MDCT can help in obtaining detailed data on the morphology of the heart, great vessels, the anatomy of the internal organs, and their mutual arrangement that make an accurate diagnosis of HS.

Patient consent for publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the editor of this journal.

CrediT authorship contribution statement

Concept - HAE; Design - HAE; Supervision - HAE; Resources and data Collection - HAE; Literature Search - AS; Writing Manuscript - HAE; Critical Review - MWN. All authors have read and approved the final manuscript.

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