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Case Report

A new variant in the heterotaxy polysplenia syndrome

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ABSTRACT

Heterotaxy is a low incidence genetic disorder of multifactorial inheritance characterized by various abnormalities in the position of organs and vessels relative to the midline of the body. It is most often associated with fatal congenital heart defects at birth. As a result, only a small percentage of patients survive into adulthood. The following clinical case presents a female adult patient with abdominal pain secondary to choledocholithiasis. During the diagnostic workup, she was diagnosed with intestinal malrotation complicated by portal vein thrombosis, which later developed into portal hypertension. In addition to several anomalies (agenesis of the inferior vena cava with direct communication of the suprahepatic veins to the right atrium, polysplenia and shortening of the pancreas), a possible variant of heterotaxy syndrome was diagnosed.

Keywords: Heterotaxy, Polysplenia, Intestinal malrotation, Agenesis of the inferior vena cava, Adults

INTRODUCTION

During normal embryologic development of the human body, most thoraco-abdominal organs develop asymmetrically and lateralized relative to the midline. The apex of the heart, spleen, stomach, and aorta are located to the left of the midline, while the liver and inferior vena cava are located to the right, a situation known as situs solitus.[1]

Heterotaxia syndrome, or situs ambiguous, is a rare condition with an incidence of 1/10,000 live births. It is characterized by anomalies of the vasculature and the position of the abdominal organs, polysplenia, and cardiac malformations (50-90%).[1] This pathology was first described in 1929 as a congenital anomaly secondary to a defect in thoraco-abdominal organ lateralization during embryogenesis.[2]

Heterotaxy syndrome also includes intestinal malrotation, a condition with an incidence of 1-200 to 1/6000 of all living newborns. The majority of cases are diagnosed within the first month of life, and approximately 90% of patients have a diagnosis within the first year of life.[3] Adult presentation is rare, accounting for only 0.2-0.5% of cases.[4] There are several complications of this pathology, but the most common are frequent episodes of intestinal obstruction and portal and/or mesenteric thrombosis. In addition, intestinal malrotation has even been reported to favor biliary stasis due to compression

of the biliary tract or anomalous displacement of the duodenum. The treatment of intestinal malrotation focuses on the management and its complications.

CASE REPORT

A 35-year-old female with a history of cholecystectomy (2003) and appendectomy (2004) was evaluated for jaundice and abdominal pain in the right hypochondrium associated with a cholestatic pattern. An ultrasound was requested and a diagnosis of primary choledocholithiasis was made. Endoscopic retrograde cholangiopancreatography (ERCP) was performed with difficult access to the duodenal papilla due to altered anatomy. Complete extraction of the stone was not archived, requiring the placement of a biliary stent as a palliative biliary drainage (Figure 1). Despite this, she continued with abdominal pain and distention, so a contrast-enhanced abdominal tomography was requested. The test showed small bowel loops mostly located to the right of the midline, with the cecum and large bowel located to the left of the abdomen. Also, data suggestive of portal hypertension was noted (collateral vessels, splenomegaly, and portal cavernomatosis). A diagnosis of intestinal malrotation complicated with portal venous thrombosis was made. Other anatomical anomalies were also identified: agenesis of the inferior vena cava with direct communication of the suprahepatic veins to the right atrium, polysplenia, pancreatic shortening and intestinal malrotation (Figures 2 and 3). The syndrome of polysplenia heterotaxia syndrome was stablished. Endoscopic follow-up revealed a filling defect at the junction of the distal common bile duct and the common hepatic duct. Sphincteroplasty and balloon dilatation to 12 mm were performed, allowing the extraction of the stone.

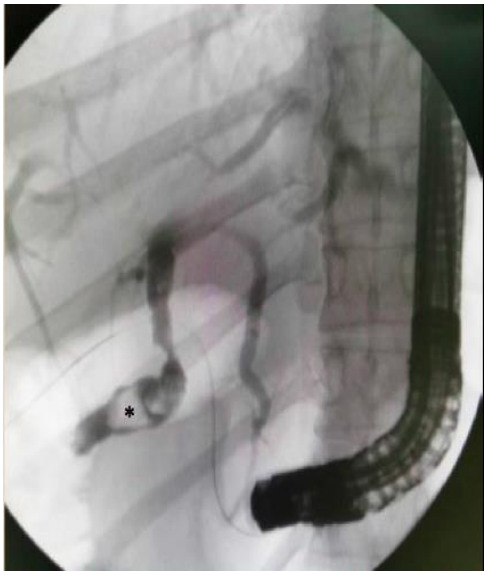

Figure 1: Cholangiography: multiple filling defects are observed, the largest being 20 mm (asterisk); anomalous arrangement of the biliary tract due to intestinal malrotation is observed.

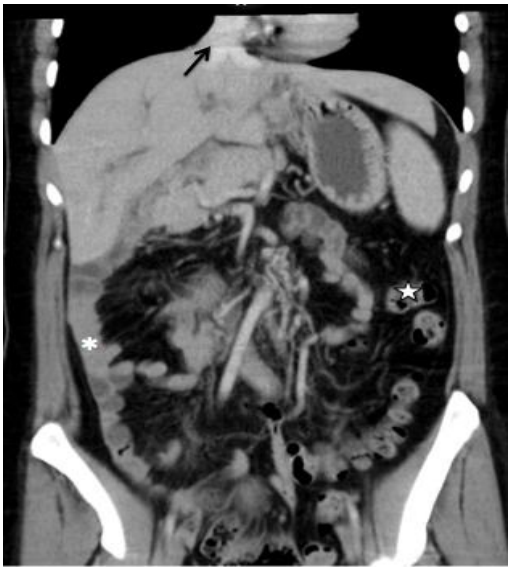

Figure 2: Contrast abdominal CT (venous phase), coronal view, showing direct drainage of the suprahepatic veins into the right atrium (black arrow); it also shows the arrangement of the intestinal loops of the small intestine in the right hemiabdomen (asterisk) and the large intestine in the left hemiabdomen (star).

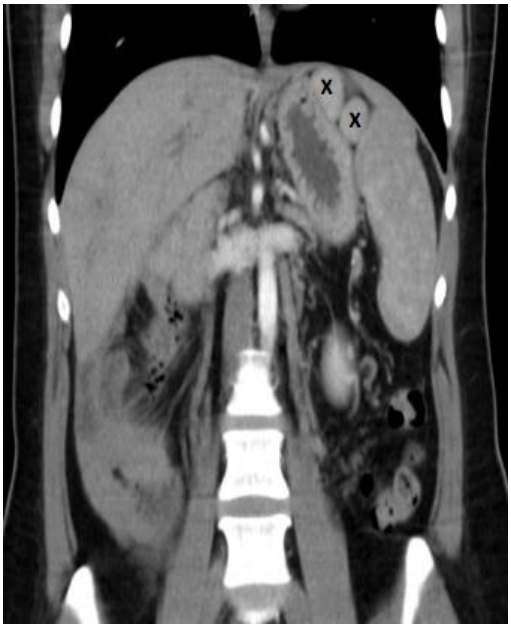

Figure 3: Contrast abdominal CT (arterial phase), coronal section, showing splenomegaly and the presence of two accessory spleens (X) of 21 and 25 mm diameter (polysplenia).

Currently, she remains asymptomatic with laboratory studies within normal parameters and medical management based on oral anticoagulants and non-selective beta-blockers for the presence of portal thrombosis and esophageal varices secondary to portal hypertension.

Table 1: Morphological characteristics of heterotaxia syndrome.[16]

Right isomerism	Left isomerism
Cardiovascular malformations	
Common atrium with bilateral right atrial appendages	Bilateral left atrial appendages
Mesocardia/dextrocardia	Complete/incomplete AV septal defect
Atrioventricular discordance	Unbalanced ventricles
Single right ventricle	Persistent left superior vena cava sometimes draining into the left atrium
AV septal defect	Interrupted hepatic portion of the inferior vena cava
Common AV connection associated with AV valve regurgitation	Partial anomalous pulmonary venous drainage
Double-outlet right ventricle	Hypoplastic sinus node (sick sinus syndrome)
Malposition of the great arteries	Single/paired AV nodes
Pulmonary stenosis or atresia	Interruption between AV node and his bundles (congenital AV block)
Total anomalous pulmonary venous drainage (with/without PVO)	
Right aortic arch	
Bilateral superior vena cava	
Bilateral sinus node	
Paired (anterior/posterior) AV nodes with sling formation	
Extracardiac malformations / Dysfunctions	
Bilateral right-sided lungs and bronchi	Bilateral left-sided lungs and bronchi
Asplenia (susceptibility to Streptococcus pneumoniae)	Bilateral hyparterial bronchi
Symmetrical liver	Polyesplenia
Right-sided stomach	Midline liver
Malrotation of the intestine	Extrahepatic biliary atresia/hypoplasia
Bronchial cilia dysfunction	Extrahepatic portal vein atresia

AV: atrioventricular; PVO: pulmonary venous obstruction.

DISCUSSION

The word heterotaxia is derived from the Greek words heteros (different) and cabs (disposition), thus this entity encompasses a wide spectrum of anomalies involving malposition and dysmorphism of the thoracoabdominal organs and vessels along the left-right axis of the body.[6,7]

Its etiology is unknown, although it is thought to result from a mutation in one of the genes necessary for normal asymmetrical left-right organ development.[8] Several inheritance mechanisms have been proposed: autosomal dominant, recessive, and X-linked recessive.[9]

This entity has been classified into two major groups: heterotaxya associated with polysplenia and heterotaxya with asplenia.[10] The incidence of polysplenia is 1/10000 live births, while the incidence of asplenia is 1/20000 live births. The variant associated with polysplenia is the most common and presents with multiple accessory spleens of variable size, usually located adjacent to the stomach (greater curvature). It is typically diagnosed in early childhood, as most of them are associated with severe cardiac anomalies in up to 50-90% of the cases, resulting in death of the patients before the age of 5 years.[11,12] Only 5-10% of patients with polysplenia syndrome have normal hearts or minor cardiac defects that allow them to reach adulthood without symptoms.[8] Therefore, the diagnosis in these patients is usually made incidentally during imaging studies or vascular procedures for other reasons.[13]

Patients with heterotaxya syndrome have also been subdivided into right-sided isomerism or left-sided isomerism (more common) according to the characteristic morphology of the atrial appendages of the heart. The word isomerism is derived from the Greek: iso (equal) and meros (part) and is used to refer to structures whose two parts are mirror images of their contralateral sides.[14] The most characteristic anomalies in this syndrome are bilateral bi-lobulated lungs, bilateral pulmonary atria, polysplenia, and interruption of the inferior vena cava with continuation of the azygos or hemi-azygos vein (Table 1).[12,15,16]

Other described anomalies include intestinal malrotation (60% of cases), hepatic drainage directly into the cardiac cavities, short pancreas, preduodenal portal vein, changes in the position of the liver (50%), and gallbladder. Abnormalities of the biliary tract include biliary atresia (neonates), and gallbladder hypoplasia or agenesis (infants). Biliary abnormalities in adults are rare, with

only 5-10% of patients with heterotaxya with polysplenia surviving into adolescence.[1]

The most common vascular anomalies are the interruption of the inferior vena cava above the origin of the renal veins, with prolongation through the azygos or hemi-azygos vein (the most common anatomical condition), and hepatic drainage directly into the cardiac cavities through a common trunk, which is usually unique.[17] Other relevant findings include the presence of a preduodenal portal vein (50%), which may interfere with the normal development of the pancreas, causing anomalies such as an annular pancreas; however, the most common anomaly is the short pancreas.[18]

A higher incidence of neoplasia, such as gastrointestinal tumors (stomach, gallbladder, pancreatic, and colorectal cancers), has been reported in adults with heterotaxia syndrome with polysplenia and in patients with intestinal malrotation.[19]

It is interesting how this case initially presented with a picture of primary choledocholithiasis, probably favored by intestinal malrotation, which was later associated with other anatomical alterations that finally contributed to the diagnosis of a possible variant of heterotaxya with characteristics of both types of isomerism.

CONCLUSION

Heterotaxia syndrome with congenital polysplenia is a rare disorder in adult patients that requires a high index of suspicion because there is no single pathognomonic alteration and its diagnosis is based on a set of multiple abnormalities. Therefore, a multidisciplinary approach should be adopted for its diagnosis, management and follow-up. Although congenital, it is important to consider this pathology in the differential diagnosis of adult patients.

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REFERENCES

1. Martínez CP, Vila Costasa JJ, Sanchotenab JL. Síndrome de heterotaxia (poliesplenia) en un adulto. Radiología. 2006;49(3):211-4.
2. Marton T, Cesko I, Hajdu J, Hargitai B, Papp Z. Heterotaxy syndrome, analysis of 13 cases and review of the literature. Orv Hetil. 2002;143:299-301.
3. Kapfer AS, Rappold FJ. Malrotación intestinal: no es sólo un problema del cirujano pediátrico. Revista del Colegio Americano de Cirujanos. 2004;199(4):628-35.
4. Butterworth WA, Butterworth WJ. Una presentación en adultos de vólvulo del intestino medio secundario a una malrotación intestinal: informe de un caso y revisión de la literatura. Revista Internacional De Informes De Casos De Cirugía. 2018;50:46-9.
5. Melkonian V, Quadri P, Varma CR, Nazzal M, Randall HB, Nguyen MJP. asymptomatic intestinal malrotation progressing to midgut volvulus in a decompensated alcoholic cirrhotic adult: a rare scenario requiring special considerations. Case Rep Surg. 2020;2020:4196012.
6. Andeson RH, Baker EJ, Penny DJ, Redington AN, Rigby ML, Wernovsky G. Pediatric Cardiology. 3rd ed. Philadelphia: Churchill Livingstone/Elsevier; 2010.
7. Jacobs JP, Anderson RH, Weinberg PM. The nomenclature, definition and classification of cardiac structures in the setting of heterotaxy. Cardiol Young. 2007;17(2):1-28.
8. Winer-Muram HT, Tonkin ILD. The spectrum of heterotaxic syndromes. Radiol Clin North Am. 1989;27:1147-70.
9. Gagner M, Munson L, Scholz FJ. Hepatobiliary anormalies associated with polysplenia síndrome. Gastrointest Radiol. 1991;16:167-71.
10. Fulcher AS, Turner MA. Abdominal manifestations of situs anomalies in adults. Radio Graphics. 2002;22:1439-56.
11. Kayhan A, Lakadamyali H, Oommen J. Polysplenia syndrome accompanied with situs inversus totalis and annular pancreas in an elderly patient. Clin Imaging. 2010;34:472-5.
12. Gayer G, Apter S, Jonas T. Polysplenia syndrome detected in adulthood: report of eight cases and review of the literature. Abdom Imaging. 1999;24:178-84.
13. Vijayvergiya R, Bhat MN, Kumar RM. Azygos continuation of interrupted inferior vena cava in association with sick sinus syndrome. Heart. 2005;91:26.
14. Kim SJ. Heterotaxy syndrome. Korean Circ J. 2011;41(5):227-32.
15. Malki MC, Outznit M, Mechhor S. Polysplenia syndrome in adulthood: a case report. Pan Afr Med J. 2022:41-67.
16. Shiraishi I, Ichikawa H. human heterotaxy syndrome from molecular geneticts to clinical features, management, and prognosis. Circul J. 2012;76.
17. Gomez A, Sanjuan JC, Martín D, Diego J, Serrano MA. Poliesplenia con heterotaxia visceral abdominal parcial y páncreas pequeño. Rev Esp Enfer Dig. 1994;85:395-7.
18. Muñoz JJ, Ramirez D, Anthon FJ, Podgaetz E, Ávila Flores F, Chan C. Polysplenia síndrome in the adult patient. Case report with review of the literatura. Ann Hepatol. 2004;3:114-7.
19. Dimpankar R, Morimoto M. Malrotation of the intestine in adult and colorrectal cancer. Review Article. Indian J Surg. 2015;77(6):525-31.

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