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Images of Interest / Imagens de Interesse

Left Isomerism - Magnetic Resonance Diagnosis

Síndrome Heterotáxica com Poliesplenia – Diagnóstico Tomográfico

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Abstract

Heterotaxy syndrome is a rare congenital condition with multifactorial inheritance in which the internal organs are abnormally arranged in the chest and abdomen. It can be classified into two main subcategories: heterotaxy syndrome with polysplenia (left isomerism) and heterotaxy syndrome with asplenia (right isomerism). Such a distinction between these subcategories offers better means of indicating the genetic alterations responsible for the syndrome. Individuals with this condition have complex congenital abnormalities, affecting various organs such as the heart - interferes in blood transport - and lung - altering the number of pulmonary lobes. We report a case of heterotaxy syndrome with polysplenia without abnormality in the thoracic organs in an adult patient.

Keywords

Heterotaxic syndrome; Polysplenia; Agenesis of the body and tail of the pancreas; Magnetic resonance imaging; Diagnosis.

Resumo

A síndrome heterotáxica é uma condição congênita rara com herança multifatorial em que os órgãos internos estão arranjados anormalmente no tórax e abdome, podendo ser classificadas em duas subcategorias principais: síndrome heterotáxica com poliesplenia (isomerismo esquerdo) e síndrome heterotáxica com asplenia (isomerismo direito). Indivíduos com esta condição têm complexas anormalidades congênitas acometendo diversos órgãos, como o coração – dificultando o transporte sanguíneo – e o pulmão – alterando o número de lobos. Relatamos um caso de síndrome heterotáxica com poliesplenia sem anormalidade nos órgãos torácicos em paciente adulta.

Palavras-chave

Síndrome heterotáxica; Poliesplenia; Agenesia do corpo e cauda do pâncreas; Ressonância Magnética; Diagnóstico.

Introduction

The heterotaxy syndrome is a rare congenital anomaly, with a high mortality rate due to the multiple cardiac abnormalities that may be classified in two major subcategories: heterotaxy syndrome with polysplenia (left isomerism) and heterotaxy syndrome with asplenia (right isomerism).[1,2,3] The polysplenia subcategory is associated with multiple spleens, in contrast to the asplenia subcategory.[1,2] However, both involve a large number of findings, which often overlap.[1,2]

Heterotaxy syndrome with asplenia is associated with congenital heart disease in 99–100% of the cases and is usually more severe compared to heterotaxy syndrome with polysplenia.[1,2] This explains the higher frequency of diagnoses of heterotaxy syndrome with polysplenia made incidentally in adulthood since the average lifespan of those with cardiac abnormalities is up to the first decade of life.[1,2] The 5-year survival of right isomerism is around 30% – 74%.[4]

Left isomerism has a less complex associated cardiac defect, which can be managed better, with the survival rate around 65% and 84%.[4] Both right and left isomerism can present arrhythmias.[5] The main characteristics of both isomerism are demonstrated in table 1.[4,6] Both isomerisms can present agenesis of the corpus callosum, central nevous system abnormalities – spina bifida, clef plate, and intestinal malrotation.[6]

We demonstrate a case of heterotaxy syndrome with polysplenia without abnormality of the thoracic organs in an adult patient.

Table 1 – Summary of right and left isomerism characteristics.

Characteristics	Right isomerism	Left isomerism
Cardiovascular malformations	Atrioventricular discordance	Bilateral left atrial appendages
	Bilateral superior vena cava	Interrupted hepatic portion of inferior vena cava
	Double-outlet right ventricle	Left superior vena cava
	Malposition of the great arteries	Partial anomalous pulmonary venous drainage
	Mesocardia/dextrocardia	Persistent left inferior vena cava draining into the left atrium
	Pulmonary stenosis/atresia	
	Right-sided aortic arch	
	Single atrium with bilateral right atrial appendages	
	Single right ventricle	
	Total pulmonary venous drainage	
Other malformations	Asplenia	Bilateral left-sided lungs and bronchi
	Bilateral right−sided lungs and bronchi	Bilobed lungs
	Right-sided stomach	Extra-hepatic biliary atresia/hypoplasia
	Short bronchus	Extra-hepatic portal vein atresia
	Trilobed lungs	Long bronchus.
		Polysplenia

Case Report

A 32-year-old female patient with longstanding abdominal pain. She mentions two previous cesarean sections. She presents a previous ultrasound examination describing a

suspicion of a right adrenal gland nodule. The MRI shows ectopic spleen tissue simulating a right adrenal nodule; intestinal malrotation with gastric fundus on the right; dorsal pancreas agenesis. However, the heart is in its usual position in the left hemithorax (Figures 1, 2, and 3). The set of findings is compatible with heterotaxy syndrome with polysplenia without abnormalities in the thoracic organs. The patient is now under outpatient follow-up as the patient was asymptomatic and the MRI did not show volvulus or obstruction from a mesenteric band.

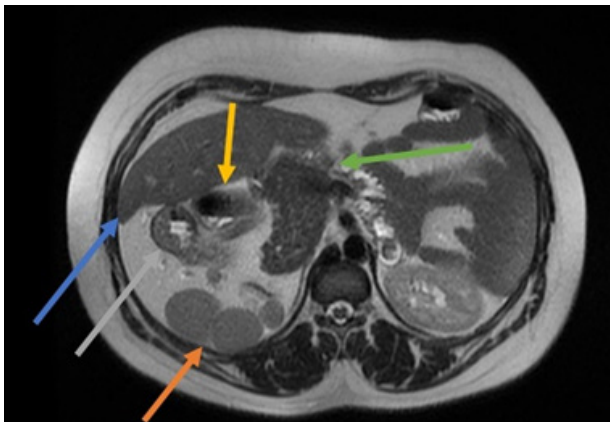

Figure 1 – MRI in axial section in T2 sequence showing usual hepatic parenchyma in the right abdomen (blue arrow), ectopic splenic tissue with splenosis (orange arrow), gastric curvature in the right abdomen (gray arrow), duodenum (yellow arrow), and dorsal pancreas agenesis (green arrow).

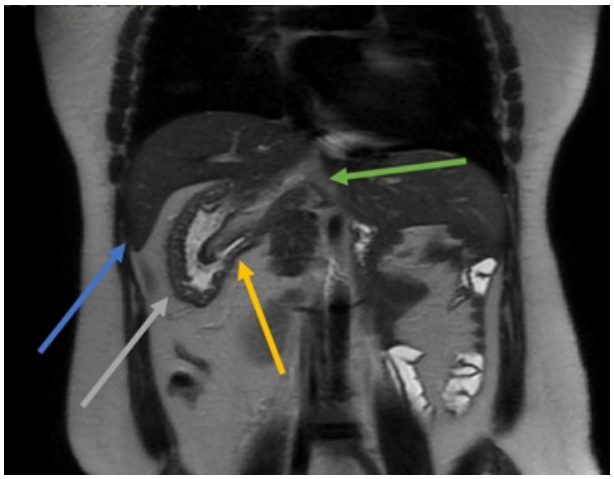

Figure 2 – MRI in the coronal section of the T2 sequence showing usual hepatic parenchyma in the right abdomen (blue arrow), ectopic splenic tissue with splenosis (yellow arrow), gastric curvature in the right abdomen (gray arrow), and dorsal pancreas agenesis (green arrow).

Discussion

The orderly and habitual arrangement of organs in the human body begins with embryonic formation, based on genetic data and the loss of this organization can characterize situs inversus or a disordered and variable arrangement, as in the case of heterotaxy syndrome.[7] The disease is more prevalent in men (2 male: 1 female).[7]

Although the heterotaxy syndrome with polysplenia and pancreatic disorder is a rare anomaly, it can be associated with the increased risk of pancreatitis and diabetes mellitus.8 Dorsal pancreatic agenesis is mostly encountered incidentally in an asymptomatic patient.[8] This finding occurs when searching for other diseases.[8] Nevertheless,

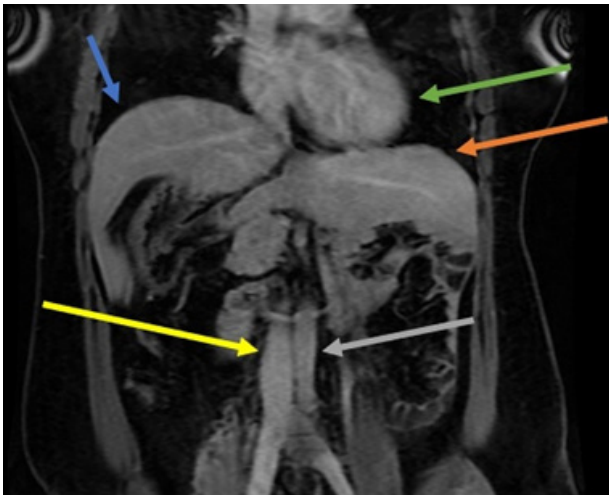

Figure 3 – MRI in the coronal section of the T1 FAT SAT sequence with contrast showing right hepatic lobe (blue arrow), left hepatic lobe (orange arrow), aorta (gray arrow), inferior vena cava (yellow arrow), and left heart ventricle (green arrow).

patients can complain of abdominal pain and present hyperglycemia which is noticed in about 50% of the cases - it happens because of the loss of islet cells that are mostly located in the tail and body of the pancreas.[8]

Although heterotaxy syndrome with polysplenia is related to multiple spleens in most patients, some patients may present with only a lobulated spleen or even a normal spleen and may be associated with congenital heart disease, interruption of the inferior vena cava (IVC) with continuation to the azygous system, intestinal malrotation, pelvic and pancreatic abnormalities.[1,2]

In normal embryologic development, during the 6th week of fetal life, the midgut herniates into the umbilical stalk.9 It also undergoes a 90-degree counterclockwise rotation around the superior mesenteric artery.[9] In the 10th week, the midgut returns to the abdomen and ends an additional 180-degree counterclockwise rotation.[9] Malrotation directs to the failure of the midgut to complete the 270-degree counterclockwise rotation around the superior mesenteric artery – an action that can be stopped at any phase, with nonrotation being one of the possible consequences. In nonrotation, the first 90-degree counterclockwise rotation is concluded. But there is no further rotation once the midgut returns to the abdominal cavity, resulting in small bowel on the right and colon to the left of the midline.[9] In general, patients are asymptomatic, and abdominal symptoms are most common in adults, and sometimes CT demonstrates intestinal malrotation.[10]

The syndrome is a rare congenital disorder, but the radiological recognition of the wide variety of presentations, as well as its adequate description, represents the best way to evaluate cases with a high risk of complications resulting from anatomical changes.[11] The patients with complex cardiac lesions have a mortality of 85% per year if they present asplenia and 50% in patients with polysplenia.[2]

The heterotaxy syndrome is a challenge and should be incorporated in a long-term treatment strategy.[2] Albeit surgical death remains the leading cause of mortality, the sudden death is also common in patients with syndrome and atrial isomerism, which is characterized by symmetrical organs which are generally asymmetric,[2] wherein, the survival of infants with isomerism is estimated at 64% at 5 years, 57% at 10 years and 53% at 15 years.[12]

The previously called "asplenia" usually presents duplication of the structures present in the right side, having trilobate lungs, left atrium with morphology equivalent to the right atrium, centrally positioned liver, aorta, and inferior vena cava located in the left, in addition to intestinal malrotation.[7] In the previously called "polysplenia" there is usually duplication of the structures of the left side, with bilobar lungs, right atrium anatomically identical to the left atrium, liver with a central position, absence of the hepatic segment of the inferior vena cava with a continuation by the azygos or hemiazygos vein, in addition to intestinal malrotation.[3] Cardiac abnormalities are less frequent and milder, explaining a higher prevalence of these findings in older individuals.[7]

Early surgical intervention is critical for those within this population who have cardiac abnormalities and gastrointestinal abnormalities, as it improves survival.[2] But, despite interventions, mortality remains high.[2] A frequent follow-up with a multidisciplinary team is of capital importance.[2] Thus, radiological evaluation is essential in identifying and planning the approach to patients with cardiac complications.[7]

Ethical disclosures / Divulgações Éticas

Conflicts of interest: The authors have no conflicts of interest to declare.

Conflitos de interesse: Os autores declaram não possuir conflitos de interesse.

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Confidentiality of data: The authors declare that they have followed the protocols of their work center on the publication of data from patients.

Confidencialidade dos dados: Os autores declaram ter seguido os protocolos do seu centro de trabalho acerca da publicação dos dados de doentes.

Protection of human and animal subjects: The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Protecção de pessoas e animais: Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pelos responsáveis da Comissão de Investigação Clínica e Ética e de acordo com a Declaração de Helsínquia da Associação Médica Mundial.

Referências

1. Gonçalves CM, Noschang J, Silva ACB, Mello RJK, Schuh SJ, Maciel AC. Heterotaxy syndrome: a case report. Radiologia Brasileira. 2014;47:54-6.
2. Lagrotta G, Moises M. Heterotaxy polysplenia syndrome in adulthood: focused review and a case report. Cureus. 2020;12:e6822.
3. Loomba RS, Hlavacek AM, Spicer DE, Anderson RH. Isomerism or heterotaxy: which term leads to better understanding? Cardiol Young. 2015;25:1037-43.
4. Huang Lili, Mitchell Belinda J, Andronikou Savvas, Lockhat Zarina I, Suleman Farhana. Heterotaxy syndrome: this is the left, right? Journal of Radiology. 2015;19:1-5.
5. Loomba R, Shah PH, Anderson RH, Arora Y. Radiologic considerations in heterotaxy: the need for detailed anatomic evaluation. Cureus. 2016;8:e470.
6. Thomas C, Sawyer SN. Prenatal findings in left atrial isomerism and an overview of heterotaxy syndrome. Journal of Diagnostic Medical Sonography. 2018;34:148-54.
7. Carneiro DS, Arantes JH, Souza GV, Barreto AS, Cardoso ML, Gontijo F. Heterotaxy syndrome: a case report. Radiologia Brasileira. 2013;46:181-3.
8. Riguetto CM, Pelichek S, Moura Neto A. Heterotaxy syndrome with agenesis of dorsal pancreas and diabetes mellitus: case report and review of the literature. Arch Endocrinol Metab. 2019;63:445-8.
9. Pagkratis S, Kryeziu S, Lin M, Hoque S, Bucobo JC, Buscaglia JM, Georgakis GV, Sasson AR, Kim J. Case report of intestinal non-rotation, heterotaxy, and polysplenia in a patient with pancreatic cancer. Medicine (Baltimore). 2017;96:e8599.
10. Jeong JH, Kim GH, Song GA, Lee DG, Moon JY, Cheong JH, Kim S. Polysplenia syndrome with congenital agenesis of dorsal pancreas presenting as acute pancreatitis and the role of endoscopic ultrasonography in its diagnosis. Korean J Gastroenterol. 2012;60:47-51.
11. Wu MH, Wang JK, Chiu SN, Tseng WC, Lu CW, Lin HC, Lin MT, Chen CA. Twin atrioventricular nodes, arrhythmias, and survival in pediatric and adult patients with heterotaxy syndrome. Heart Rhythm. 2021;18:605-12.
12. Jaramillo A, Duaque MA, Buitrago M, Espinel A, Torres C. Síndromes de heterotaxia presentación de caso y revisión de la literatura. Revista Médica Clínica del Country. 2017;7:19-22.