

Heterotaxy syndrome associated with Polysplenia (left isomerism). About a case

Síndrome de heterotaxia asociado a poliesplenia (isomerismo izquierdo). A propósito de un caso

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Palabras clave (DeCS)
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Situs ambiguus
Disnea

Summary
The heterotaxy syndrome associated with polysplenia (left isomerism) is part of what is known as Heterotaxia or *Situs Ambiguos,* where a random location of the thoraco-abdominal organs is observed with or without vital functional compromise. We present the case of an 80-year-old male patient, with a history of high blood pressure and type 2 diabetes mellitus, who was admitted for dyspnea, weakness, and abdominal pain. During his stay, he progresses to multiple organ failure and dies. Among the routine studies, computed tomography of the chest and abdomen showed a series of anatomical alterations.

Resumen
El síndrome de heterotaxia asociado a poliesplenia (isomerismo izquierdo) hace parte de lo que se conoce como heterotaxia o *Situs Ambiguos*, en el que se observa una ubicación aleatoria de los *órganos* toracoabdominales con o sin compromiso funcional vital. Se presenta el caso de un paciente masculino de 80 años de edad, con antecedente de hipertensión arterial y diabetes *mellitus* tipo 2, quien ingresa por disnea, debilidad y dolor abdominal. Durante su estancia progresa a falla multiorgánica y fallece. Entre los estudios de rigor, la tomografía de tórax y abdomen permiten evidenciar una serie de alteraciones anatómicas.

Introduction
Heterotaxia syndrome or *Situs Ambiguous* is a rare genetic disorder with an incidence of about 1 per 10,000 live births and an accentuated prevalence in Asians (1). It is characterized by alterations in the position of the organs and vasculature associated with morphological changes in the thoracoabdominal organs, mainly the heart, lungs and spleen (2). There are two known expressions or variants of heterotaxia, which are related to the spleen, an asplenic form or right isomerism, and polysplenia, or left isomerism (3), which can go beyond the only aberrant anatomical description, with functional and vital implications in the affected persons.

Case
An 80-year-old male patient with a history of arterial hypertension (AHT) and type 2 diabetes mellitus was admitted to the emergency department for dyspnea, weakness and abdominal pain that had worsened in the last few hours. On physical examination, the patient presented marked mucocutaneous pallor, tachycardia, tachypnea and desaturation, for which reason computed tomography (CT) of the chest, abdomen and pelvis with oral contrast medium was performed. The images showed multiple thoracic and abdominal anatomical abnormalities including: hypoarterial bronchi (Figure 1), interrupted and absent inferior vena cava (IVC) in the liver, azygos-hemiazygos vein draining into the superior vena cava (SVC) (Figures 2 and 3), bridged (medialized) liver and central gallbladder (Figure 4), multiple splenules (polysplenia) (Figure 5), intestinal malrotation (Figure 6), duodenum and proximal jejunum in antero-central location (Figure 7), truncated (short) and incomplete annular pancreas (Figure 8), absence of the celiac trunk (Figure 9), and left bronchial isomerism (Figure 10). Complementary paraclinical examinations showed increased creatinine, arterial gases with metabolic acidosis, electrocardiogram with atrial extrasystoles, right bundle branch block, transthoracic echocardiogram with findings of mitroaortic valvulosclerosis, severe concentric non-obstructive mixed hypertrophy of the left ventricle with LVEF of 60%, with no anatomical alterations. The patient was admitted to the intensive care unit (ICU) and died due to multiorgan dysfunction.

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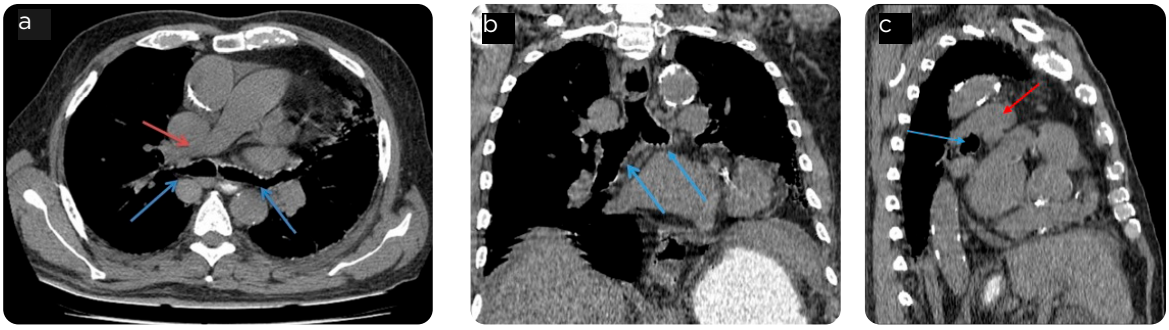


Figure 1. Simple chest CT. a) Axial section; b) coronal, and c) sagittal, in mediastinal window. Hyparterial bronchi (blue arrows), right pulmonary arterial trunk superior to the bronchi (red arrow in a) and both branches of the pulmonary arterial trunk (red arrow in c).

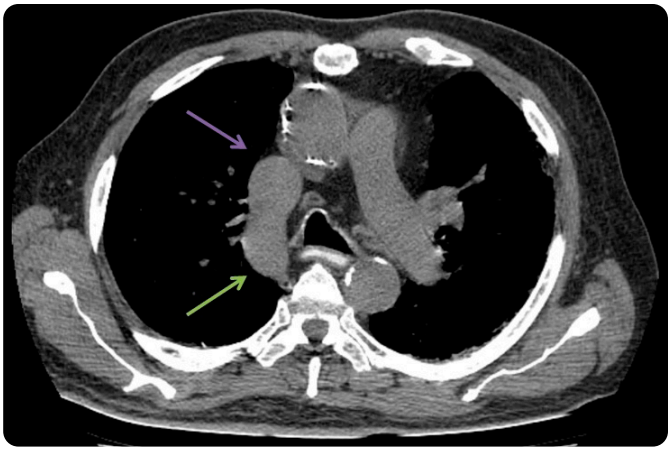


Figure 2. Simple chest CT in axial section and mediastinal window. Azygos vein (green arrow) draining into superior vena cava (purple arrow).

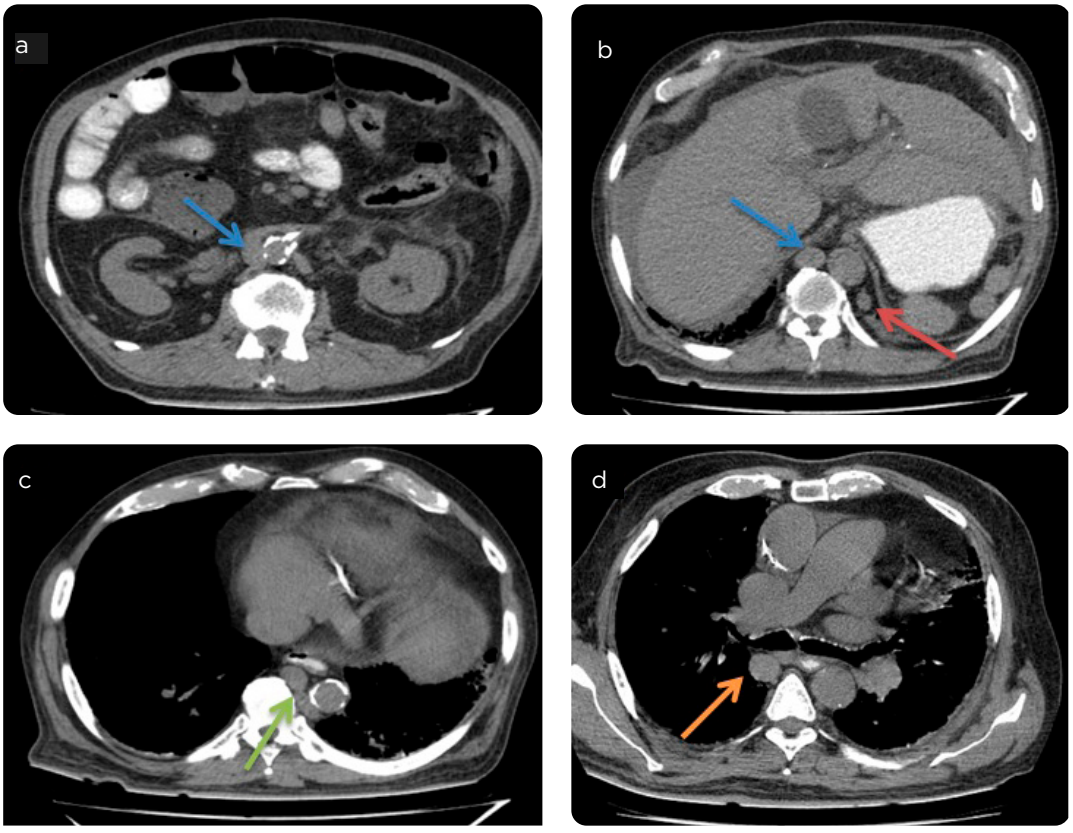


Figure 3. CT of abdomen and pelvis with oral contrast medium, axial sections and extension to both pulmonary bases. a) Left renal vein draining into the IVC (blue arrow). b) In the suprarenal sections, the IVC is interrupted and continues with the azygos vein (blue arrow), joining the hemi-azygos vein (green arrow in c and red arrow in b). The azygos vein continues at thoracic level (orange arrow in d) until it drains into the SVC as shown in figure 2. Additional findings: right subphrenic and perirenal free fluid.

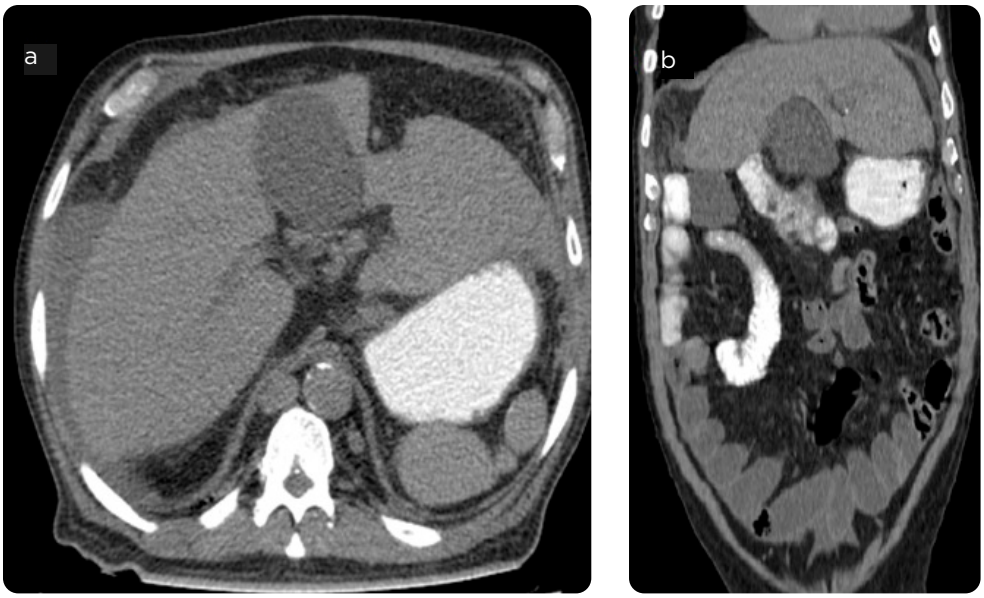


Figure 4. Abdominal and pelvis CT with oral contrast medium in axial (a) and coronal (b) sections: bridged liver (medialized) and central gallbladder.

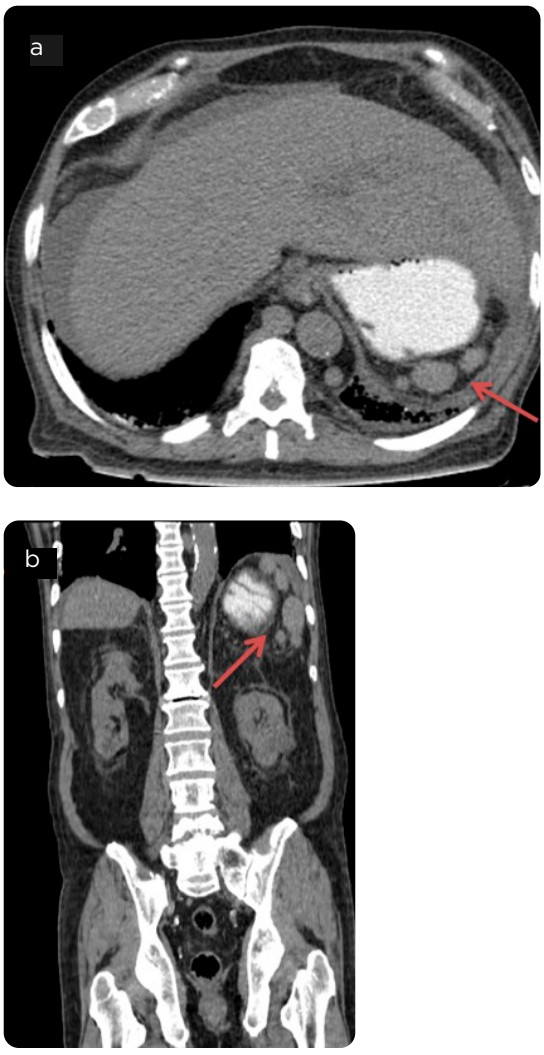


Figure 5. CT of abdomen and pelvis with oral contrast medium in axial (a) and coronal (b) slices: bridged (medialized) liver and central gallbladder.



Figure 6. CT of abdomen and pelvis with oral contrast medium, axial (a) and coronal (b) sections: multiple splenules (polysplenia) (red arrows). coronal. Signs of intestinal malrotation with thin intestinal loops located in the right hemiabdomen (purple arrow), and colon loops in the left hemiabdomen (orange arrow).

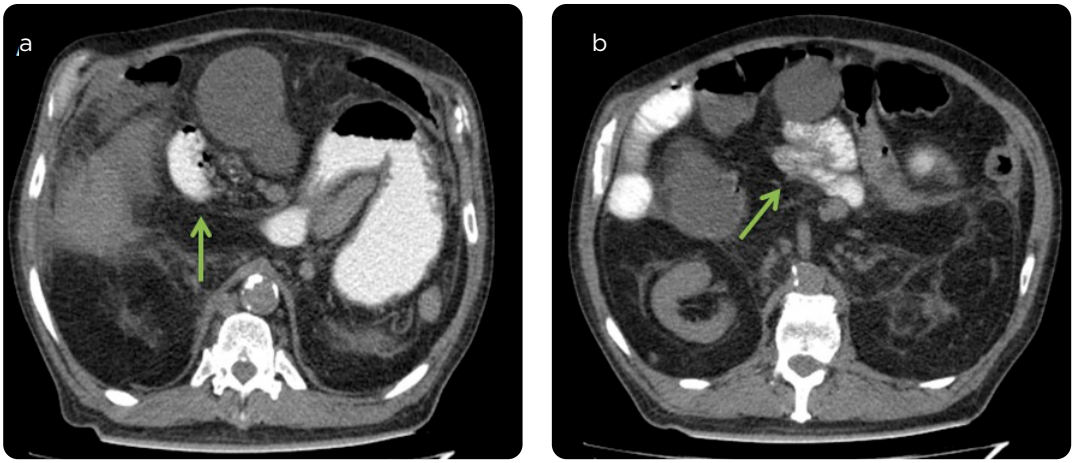


Figure 7. Abdominal and pelvis CT with oral contrast medium, axial sections at epigastric level (a) and at mesogastric level (b): alteration in the normal axis of the duodenum and proximal jejunum located anteriorly and centrally (green arrows) with narrow lateralization of the small bowel loops.

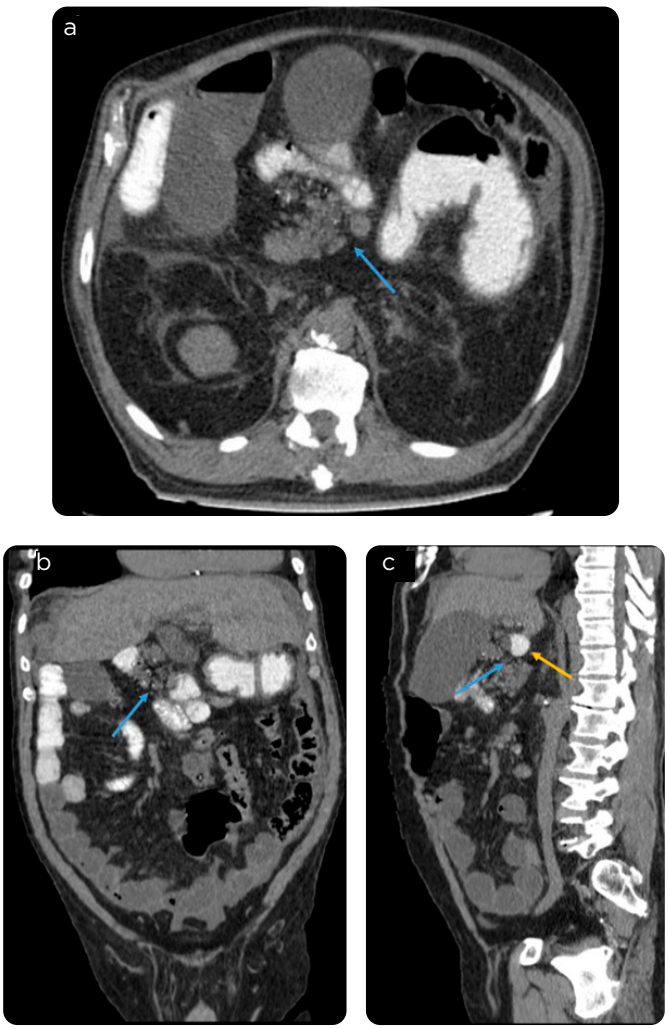


Figure 8. Abdominal and pelvis CT with oral contrast medium. a) Axial section; b) coronal, and c) sagittal. In a, truncated (short) pancreas is visualized (blue arrow). Incomplete annular pancreas indicated by blue arrows in b and c partially surrounding the duodenum indicated by yellow arrow in c.

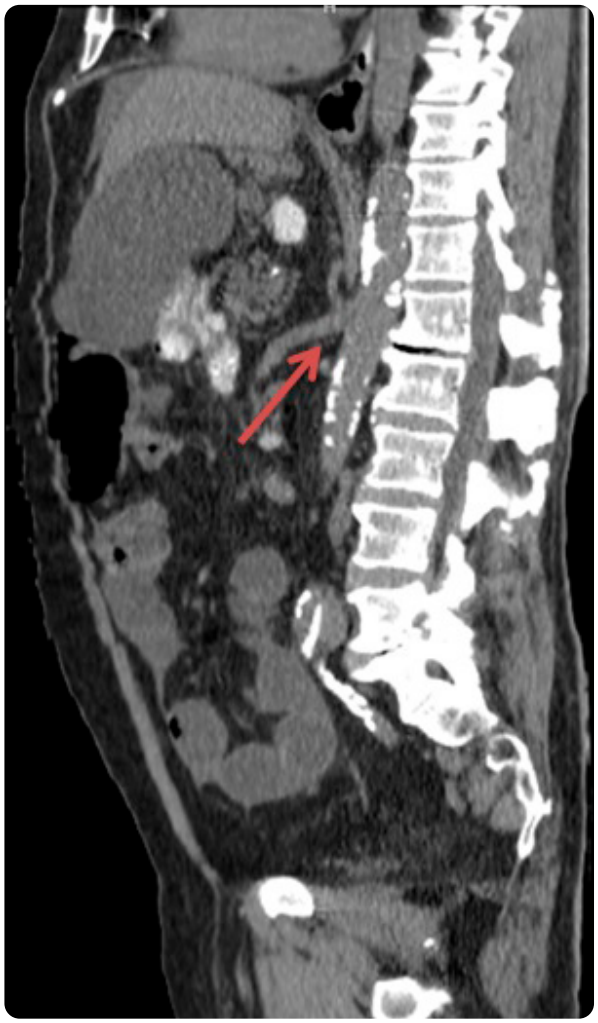


Figure 9. CT of the abdomen and pelvis with oral contrast medium, sagittal view. Absence of the celiac trunk. Superior mesenteric artery (red arrow) giving origin to a branch that is not possible to identify given the anatomical alterations described in previous figures and because there is no intravenous contrast study.

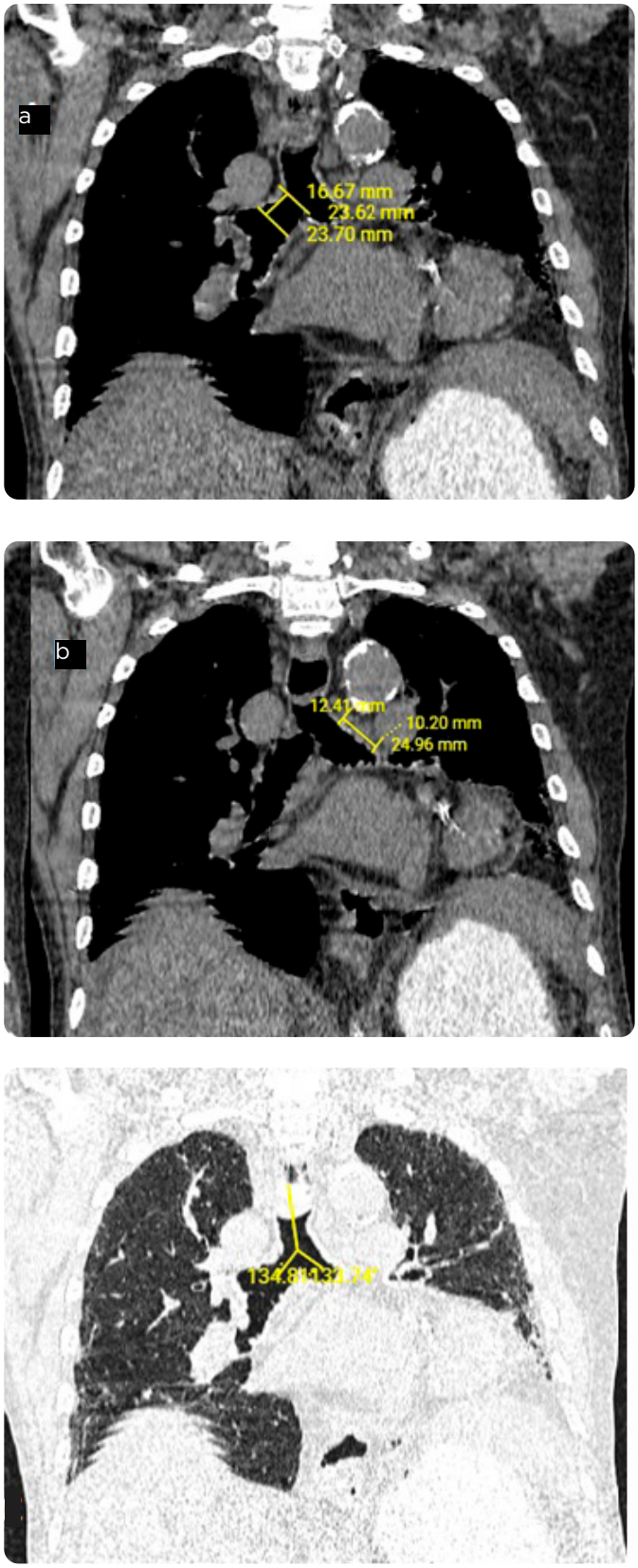


Figure 10. Simple chest CT. a and b) Coronal slices, in mediastinal window with bronchial length measurements (bronchial ratio less than 1.5). c) Pulmonary window with bronchial angle measurements (< 135°), findings related to left bronchial isomerism.

Discussion

According to the anatomical and morphological features, heterotaxy syndrome can be classified as asplenic or polysplenic variant and each of them will present certain unique or shared findings. Asplenic heterotaxia or right isomerism is generally associated with poor prognosis and the high mortality rate is due to cardiac alterations (4, 5): dextrocardia, common atrium with bilateral right atrial appendages (appendages), atrioventricular discordance, single right ventricle, double outlet right ventricle, malposition of the great arteries, pulmonary stenosis or atresia, right aortic arch, bilateral superior vena cava and bilateral sinus node, among many other findings of cardiac anomalies. Additionally, cardiac arrhythmias, trilobed lungs, asplenia, intestinal malrotation, right-sided stomach, etc., are usually associated (4).

In general terms, heterotaxia or left isomerism syndrome has less pronounced or even absent cardiac findings. It highlights defects of the interatrial septum, bilateral left atrial appendage, hypoplastic sinus node, bilobed lungs and hypoarterial bronchi, polysplenia, central liver and gallbladder, intestinal malrotation, truncated and annular pancreas, interruption of the intrahepatic inferior vena cava that continues with the azygos-hemiazygos system, and abnormalities in the central nervous system, such as myelomeningocele, porencephalic cyst, cerebellar agenesis, etc. (4, 6-8).

In recent years the approach to heterotaxy has changed. Currently it is taken into account whether it presents asplenic variant or right isomerism, or polysplenic variant or left isomerism, and has relied on the morphology of the auricular appendages (9) due to its consistency in the studies associated to bronchial isomerism. This is evaluated and determined according to the proportions of the bronchial lengths, being <1.5 indicative of the same; then it can be determined if it presents right isomerism when the bronchial angle is > 135° or left isomerism when it is < 135° (10).

Conclusion

Heterotaxy syndrome can affect multiple systems and organs, therefore radiologists must recognize and describe associated abnormalities comprehensively, which allows predicting future functional alterations and preparing the medical interventions deemed necessary.

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