Asplenia in left isomerism

Usnish Adhikari[1], Venkatesh Gurajala[1], Palanisamy Dinesh Raja[1], Anoop Ayyappan[2], Deepti Narasimhaiah[3], Arun Gopalakrishnan[1]

[1]Department of Cardiology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India, [2]Department of Imaging Sciences and Interventional Radiology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India, [3]Department of Pathology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India

ABSTRACT

Anatomical configurations where the viscero-atrial structures do not follow the usual arrangement or mirror-imaged arrangement is described conventionally as heterotaxy. Isomerism in the context of the congenitally malformed heart is a situation where some paired structures on opposite sides of the left–right axis of the body are, in morphologic terms, symmetrical mirror images of each other. It encompasses two separate entities, right and left isomerism, the former being usually associated with asplenia and the latter with polysplenia. We report herein a rare case of left isomerism that is associated with asplenia in a 4-year-old girl.

Keywords: Congenital heart disease, heterotaxy, Howell–Jolly bodies, polysplenia

INTRODUCTION

Anatomical configurations where the viscero-atrial structures do not follow the usual arrangement or mirror-imaged arrangement are described conventionally as heterotaxy. Isomerism in the context of the congenitally malformed heart is a situation where some paired structures on opposite sides of the left–right axis of the body are, in morphologic terms, symmetrical mirror images of each other. It encompasses two separate entities, right and left isomerism, the former being usually associated with asplenia and the latter with polysplenia. We report herein a rare case of left isomerism that is associated with asplenia in a 4-year-old girl.

CASE REPORT

A 4-year-old girl with a normal birth and developmental history, with a history of late neonatal sepsis and recurrent respiratory tract infections since infancy, was referred to our institute with a diagnosis of an atrial septal defect. Clinical examination revealed no cardiomegaly, normally split-second heart sound, and a brief midsystolic murmur in the pulmonary area. The oxygen saturation on room air was 98%. The electrocardiogram showed inverted p-waves in inferior leads, suggestive of ectopic low atrial rhythm [Figure 1a]. The ventricular function was normal on echocardiography, with no volume overload of the right heart and no pulmonary hypertension. The inferior caval vein was noted to be interrupted with hemiazygos continuation. However, the radiologist reported an absent spleen on ultrasound.

She underwent contrast computed tomography (CT) of the chest and abdomen, which showed bilateral bilobed lungs, hyparterial bronchi, and a large midline liver, consistent with left isomerism. Both atrial appendages demonstrated tubular configuration with a narrow base at their junctions to the atria, suggestive of left-sided morphology [Figure 1b and c]. Terminal crest could not be identified, and the pulmonary venous entry

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Address for correspondence: Dr. Arun Gopalakrishnan, Department of Cardiology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram - 695 011, Kerala, India.
E-mail: arungopalakrishnan99@gmail.com

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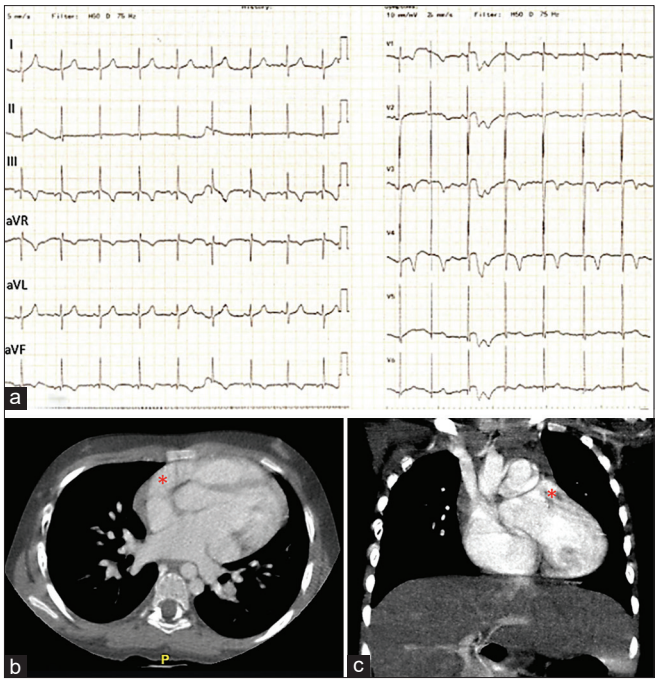

Figure 1: Panel a is the 12-lead electrocardiography showing upright P wave in I, aVL and inverted P wave in II, III, aVF (suggesting situs solitus with ectopic low atrial rhythm); normal precordial R-wave progression. Panel b and c are the contrast-enhanced computed tomography in the axial and coronal sections, respectively, demonstrating bilateral atrial appendages of left-sided morphology (red asterisks)

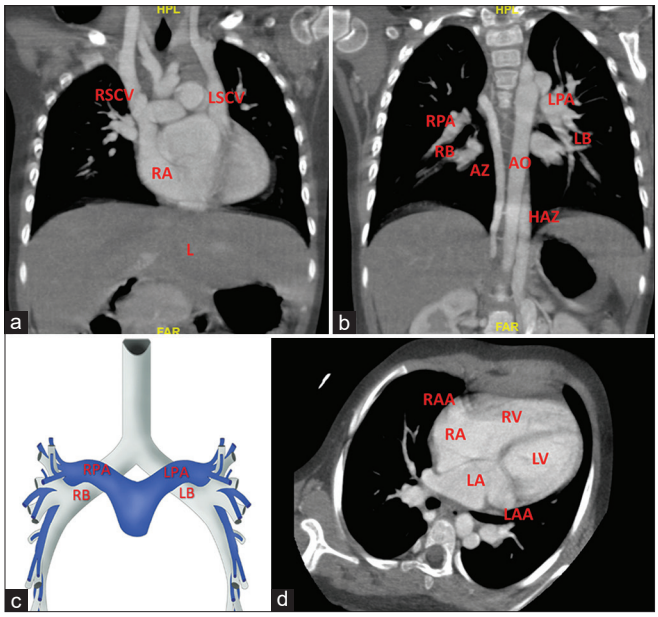

Figure 2: Clockwise from top left. The contrast-enhanced computed tomography (CT) coronal section in Panel a demonstrates the midline liver, absent spleen, bilateral superior caval veins, and absent hepatic segment of the inferior caval vein. Panel b is a posterior axial cut of the CT showing the dilated hemiazygos and azygos veins. Bilateral hyparterial bronchi consistent with left isomerism is evident in the CT in Panel 1b and the generated cartoon (Panel c). Panel d is the 4-chamber view demonstrating bilateral atrial appendages with a left-sided morphology. AO: Aorta, AZ: Azygos, HAZ: Hemiazygos, L: Liver, LA: Left atrium, LAA: Left atrial appendage, LB: Left bronchus, LPA: Left pulmonary artery, LSCV: Left superior caval vein, LV: Left ventricle, RA: Right atrium, RAA: Right atrial appendage, RB: Right bronchus, RPA: Right pulmonary artery, RSCV: Right superior caval vein, RV: Right ventricle

into the atrium was normal. The inferior caval vein was interrupted with hemiazygos continuation to a persistent left superior caval vein [Figure 2a and b] and thereafter to the dilated coronary sinus. Bilateral superior caval veins were noted. Besides the left isomeric bronchial architecture [Figure 2c], both atrial appendages demonstrated left atrial morphology [Figure 2d]. However, the CT also confirmed asplenia. The peripheral smear examination revealed a normocytic normochromic blood picture with Howell–Jolly bodies in some red blood cells [Figure 3a] and a few target cells [Figure 3b]. The white blood cells and platelets showed normal morphology. The presence of Howell–Jolly bodies in the peripheral smear further confirmed the asplenia. The child was managed with antibiotic prophylaxis as per asplenia protocol, and the restrictive atrial septal defect was considered hemodynamically insignificant.

DISCUSSION

The index case highlights the flaws of the current convention of limiting "heterotaxy" on the basis of isomerism. While isomerism refers to morphological mirror imagery of the atrial appendages (isomerism of the atrial appendages) or that of the bronchi (bronchial isomerism), heterotaxy encompasses all thoracoabdominal visceral arrangements that do not follow the usual or mirror-imaged arrangements. The extent of pectinate muscles in the atrial appendages relative to the atrioventricular junction is considered the characteristic finding to diagnose isomerism of the atrial appendages.[1,2] The presence of polysplenia or asplenia has traditionally been used as a classification parameter for heterotaxy.[1] Situs ambiguous with polysplenia has been described in the literature as bilateral left-sidedness or left isomerism, whereas in right isomerism or bilateral right-sidedness, the absent spleen is a common association. Interruption of the inferior caval vein with azygos or hemiazygos continuation of inferior veins with separated hepatic veins, bilateral hyparterial bronchi with bilobed lungs,[3] and bilateral pulmonary venous connections are frequent associations of left isomerism. However, discordance between bronchial morphology and pattern of findings in the abdominal and atrial arrangements has been documented in up to 20% of autopsy-proven heterotaxy.[4] The use of splenic anatomy to define heterotaxy can misdiagnose the type in 13% of right isomerism and 2% of left isomerism.[5] The more frequent disharmony between splenic anatomy and isomerism of the right atrial appendages, as opposed to isomerism of the left atrial appendages (25% vs. 3%–7%), was also reported by other authors.[1,6] In fact, there is more

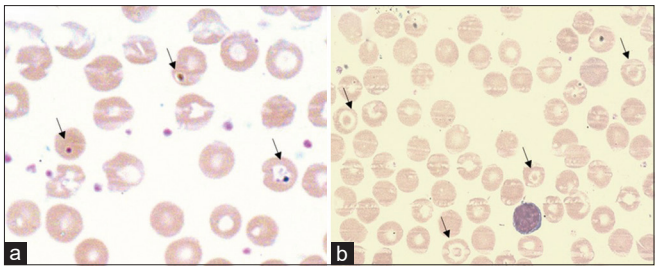

Figure 3: Peripheral smear with Howell–Jolly bodies (a, black arrows) and few target cells (b, black arrows) (Leishman stain, ×1000)

discordance when bronchial morphology is compared with splenic anatomy.[4] While the extent of disharmony led to the frequent usage of the phrase "situs ambiguous," it seems prudent to describe the anatomy in each organ system separately to simplify communication.[7] Bilateral superior caval veins are frequently associated with both forms of isomerism. Complex cardiac lesions are observed more in patients with right isomerism, which has a worse prognosis.[8] Patients with asplenia are at higher risk for severe community-acquired bacterial infections due to capsulated organisms and have a very high mortality if they develop sepsis.[9] The index patient has been on antibiotic chemoprophylaxis and is doing well at a 2-year follow-up. Our patient presents a unique case of left isomerism with asplenia, as anatomically demonstrated by imaging and with the presence of Howell–Jolly bodies on blood smear that demonstrates the fallacies of using splenic anatomy as a guide to the type of heterotaxy.

Splenic anatomy can be discordant from the bronchial and/or atrial appendage morphology and does not necessarily indicate the pattern of heterotaxy. While the atrial appendages are the only structures that may be truly isomeric, heterotaxy refers to the departure from the normal arrangement of any thoracic or abdominal visceral structure. A complete description of the individual anatomical abnormalities can remove the ambiguity in the diagnosis.

Ethical considerations

The study was conducted in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki and its amendments. Informed written consent was taken from the close relative of the patient concerned. No patient identity particulars have been disclosed.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Uemura H, Ho SY, Devine WA, Anderson RH. Analysis of visceral heterotaxy according to splenic status, appendage morphology, or both. Am J Cardiol 1995;76:846-9.
2. Anderson RH, Spicer DE, Loomba R. Is an appreciation of isomerism the key to unlocking the mysteries of the cardiac findings in heterotaxy? J Cardiovasc Dev Dis 2018;5:11.
3. Chassagnon G, Morel B, Carpentier E, Ducou Le Pointe H, Sirinelli D. Tracheobronchial branching abnormalities: Lobe-based classification scheme. Radiographics 2016;36:358-73.
4. Loomba RS, Pelech AN, Shah PH, Anderson RH. Determining bronchial morphology for the purposes of segregating so-called heterotaxy. Cardiol Young 2016;26:725-37.
5. Tremblay C, Loomba RS, Frommelt PC, Perrin D, Spicer DE, Backer C, *et al.* Segregating bodily isomerism or heterotaxy: Potential echocardiographic correlations of morphological findings. Cardiol Young 2017;27:1470-80.
6. Yim D, Nagata H, Lam CZ, Grosse-Wortmann L, Seed M, Jaeggi E, *et al.* Disharmonious patterns of heterotaxy and isomerism: How often are the classic patterns breached? Circ Cardiovasc Imaging 2018;11:e006917.
7. Mori S, Anderson RH, Nishii T, Matsumoto K, Loomba RS. Isomerism in the setting of the so-called "heterotaxy": The usefulness of computed tomographic analysis. Ann Pediatr Cardiol 2017;10:175-86.
8. Chowdhury UK, Anderson RH, Pandey NN, George N, Sankhyan LK, Khan MA, *et al.* Long-term surgical outcomes of patients with isomeric right and left atrial appendages. World J Pediatr Congenit Heart Surg 2023;14:291-9.
9. Piano Mortari E, Baban A, Cantarutti N, Bocci C, Adorisio R, Carsetti R. Heterotaxy syndrome with and without spleen: Different infection risk and management. J Allergy Clin Immunol 2017;139:1981-4.e1.