Case Report

ANNALS OF MEDICINE & SURGERY

OPEN

A rare case of inferior vena cava interruption and pulmonary valve stenosis in the absence of heterotaxy syndrome

Tuqa Rajab, MD[a], Moayad Abou Nukta, MD[b], Nour Bannoud, MD[c], Sultaneh Haddad, MD[d,e,*], Dimah Aljarmakani, MD[d], Doaa Mukhtar Mohammed Ali, MD[f], Amal Mohamed H. Hassan Jama, MD[g], Samir Srour, PhD[d]

Introduction: The complete absence hepatic portion of the inferior vena cava (IVC) is known as "interruption" of the IVC. Congenital interruption of the IVC can rarely occur in patients with a normal visceroatrial situs.
Case presentation: We present a case of an 8-year-old male child who exhibited mild dyspnea. Further evaluation revealed an interruption of the IVC and pulmonary valve stenosis, without heterotaxy.
Discussion: It is important to recognize that interruption of the IVC can occur independently or as part of a syndrome. Therefore, thorough investigations should be conducted for each patient.
Conclusion: More research is needed to explore the associations between IVC interruption and pulmonary valve stenosis.

Keywords: heterotaxy, inferior vena cava, pulmonary valve stenosis

Introduction

Patients of heterotaxy syndrome especially left isomerism typically have an azygos vein (AzV) or hemiazygos vein (HemiAzV) that continues the inferior vena cava (IVC) at the level of the liver with significant situs abnormalities.

Rarely can congenitally interrupted IVC occur in patients with a normal visceroatrial situs, and it is unclear if this condition is linked to other vascular and cardiac abnormalities.

However, the Absence of the IVC's hepatic portion was regarded as an interruption of the IVC[1].

When the hepatic portion of the IVC completely disappears, the condition is referred to as an "interruption" of the IVC. This is caused by the right subcardinal vein's inability to link with the growing hepatic veins during the seventh week of pregnancy, and it is typically linked to the supracardinal veins' persistence[1].

Only 31.3 % of cases have interruption of the IVC alone, even in the absence of other heterotaxy syndrome characteristics. In 47.8% of cases, there are additional systemic vein anomalies, such as the persistence of extra fetal venous system components or failure to form other IVC segments[1].

When an isolated interrupted IVC occurs, the patient typically shows no symptoms, and there is no need for treatment because of the vascular anomaly alone. There are cases where lower extremity venous insufficiency is observed, which might increase the risk of deep vein thrombosis in the future[2].

Ultrasound techniques can easily determine the absence of the hepatic section of the IVC with azygos continuation and distinct drainage of the hepatic veins.

The superior vena cava and the AzV exhibit a distinctive flow pattern that can be seen using Doppler radar. Magnetic resonance imaging, and computed tomography can also be used to make the diagnosis[1].

Case presentation

An 8-year-old; male child, with no significant family history, presented to the cardiac care unit with mild dyspnea, without any signs of dehydration, spasm, or convulsion. Perinatal, natal and postnatal terms were uneventful.

Physical examination revealed mild tachypnea, with no other respiratory distress signs, other vital signs were within normal limits. Jugular pulsation was visible on the right side, as well.

Upon auscultation, a systolic ejection murmur with a palpable thrill, measuring 5/6, was audible mostly at the pulmonic area, indicating a possible pulmonary valve stenosis.

The apical impulse was seen and palpable in the third intercostal space, which may indicate right ventricle hypertrophy. Additionally, it is worth noting that hepatojugular reflux test was positive, which emphasises the last findings.

Transthoracic echocardiogram (Fig. 1) exhibited normal organ and vascular positioning, with no inversion of cardiac segments, and no evidence of the presence of left superior vena cava.

Superior vena cava was draining into the right atrium, and pulmonary veins were draining into the left atrium. Furthermore, an interruption of the IVC at the level of the liver with continuation via the AzV was detected as well.

[a]Faculty of Medicine, Tartous University, Tartous, Syrian Arab Republic, [b]Faculty of Medicine, Damascus University, Damascus, Syrian Arab Republic, [c]An Ophthalmology Resident, Faculty of Medicine, Damascus University, Damascus, Syrian Arab Republic, [d]Children's University Hospital, Damascus, Syrian Arab Republic, [e]Stemosis for Scientific Research, Damascus, Syrian Arab Republic, [f]Omdurman Islamic University, Omdurman, Sudan and [g]Faculty of Medicine, Gazi University, Ankara, Turkey

Sponsorships or competing interests that may be relevant to content are disclosed at the end of this article.

*Corresponding author. Address: Children's Hospital, Damascus, Syrian Arab Republic. Tel: +963 930593703. E-mail: sltanhhdad@gmail.com (S. Haddad).

Annals of Medicine & Surgery (2025) 87:964–967

Received 24 August 2024; Accepted 14 December 2024

Published online 21 January 2025

http://dx.doi.org/10.1097/MS9.0000000000002906

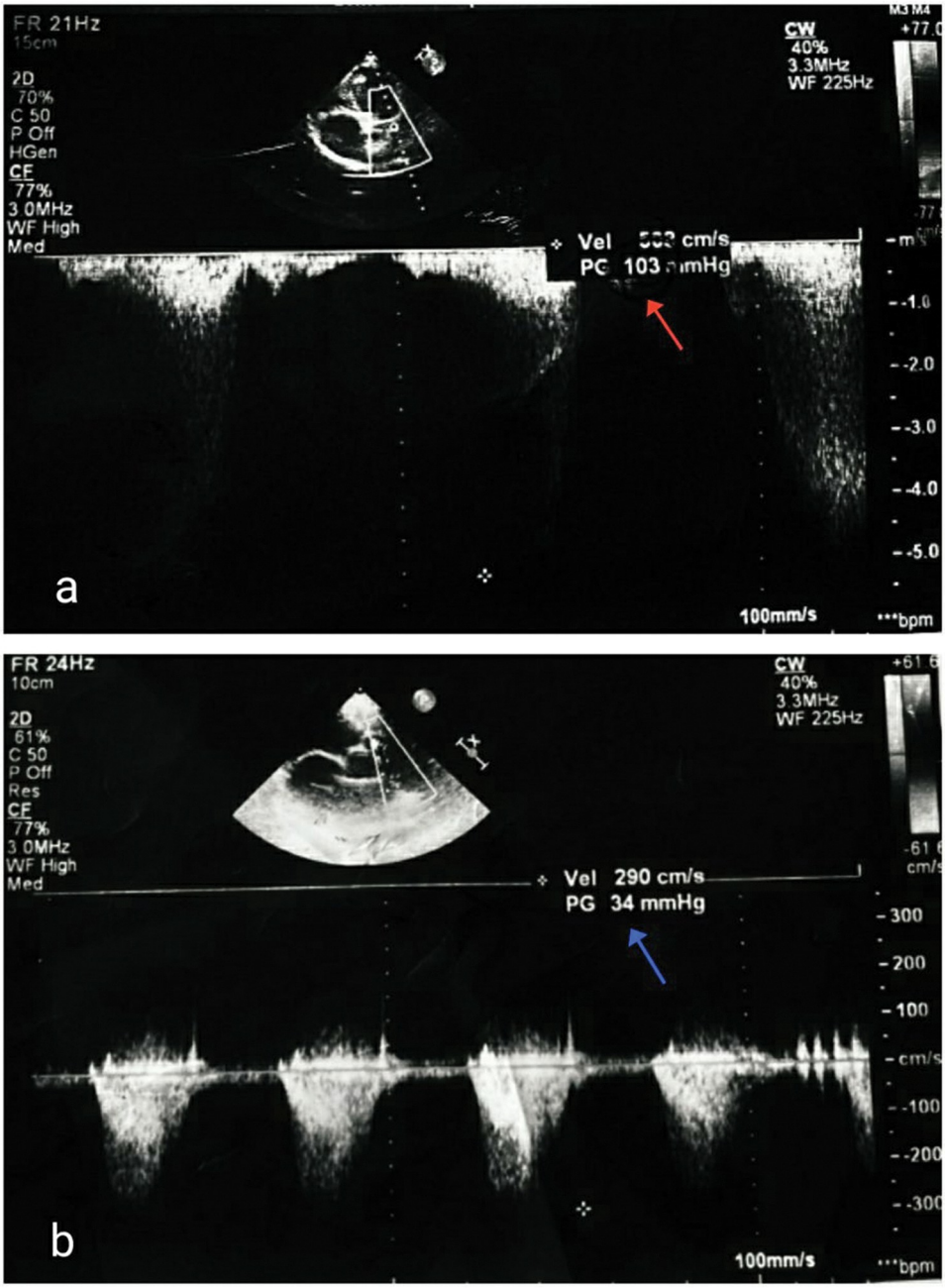

Figure 1. Transthoracic ultrasound with Doppler was used to assess the severity of pulmonary valve stenosis. Doppler assessment before (a) and after (b) pulmonary valvuloplasty showed a significant decrease in the systolic velocity through the valve, suggesting a decrease of approximately 70 mmHg in stenosis severity.

Left cardiac chambers revealed mild mitral valve regurgitation, with regular left ventricular systolic function, while the right cardiac chambers exhibited ventricular enlargement and hypertrophy, along with slight tricuspid valve regurgitation (Fig. 2). These findings suggest compensatory mechanisms for pulmonary valve stenosis, which was confirmed along with a supravalvular stenosis, where the maximum pressure gradient reached 110 mm Hg.

The abdomen ultrasound (Fig. 3) and chest radiograph excluded polysplenia, and any malposition or malformation of the lungs and abdominal organs.

Following improvements in the child's general condition, open-heart surgery was performed to repair pulmonary valve stenosis. During the procedure, the pulmonary artery was longitudinally opened above the valve to eliminate the supravalvular stenosis. Postoperative assessment revealed a notable improvement in the pressure gradient across the pulmonary valve. After 1 week of follow-up, with normal vital signs, he was discharged with no complications.

Discussion

Both pulmonary valve stenosis and interruption of the IVC are recognized as cardiac manifestations of heterotaxy syndrome[3], A rare pathological condition characterized by

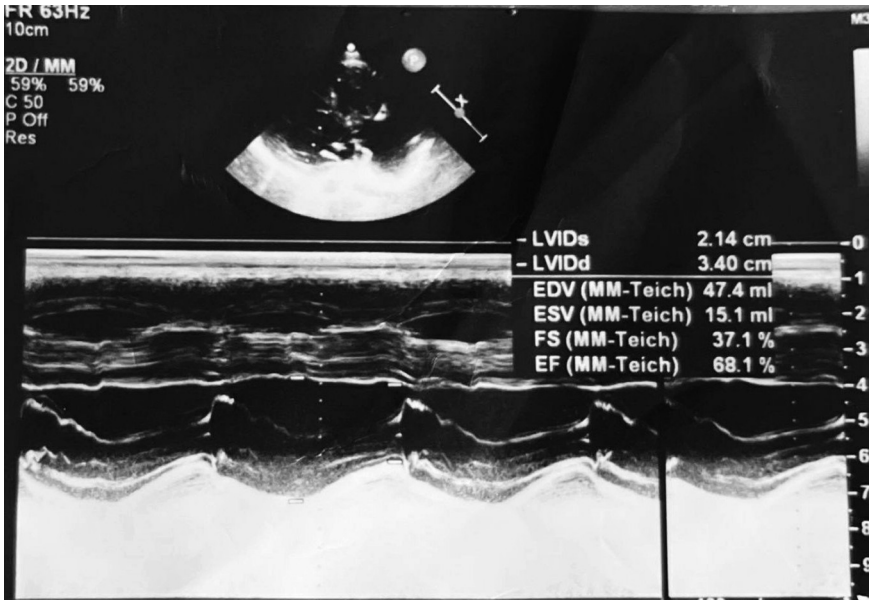

Figure 2. Echocardiographic image revealing normal left ventricular systolic function.

the atypical arrangement of internal thoracoabdominal organs across the left-right axis of the body[4], which is usually associated with polysplenia[1]. Noteworthy is the presence of situs solitus in our case, which contributes to the exceptional rarity of isolated manifestations within the spectrum of heterotaxy presentations.

Interruption of the IVC with azygos continuation arises from the failure of the subcardinal-hepatic anastomosis to form, leading to the congenital absence of the hepatic segment of the IVC. As a result, the IVC extends superiorly as a dilated azygos vein, which ultimately drains into the superior vena cava. This anomaly has an estimated prevalence of 0.6%[4]. In the case series of Bartan *et al*, associated cardiac malformations were observed in 31.3% of patients with interrupted IVC, with 4.5% presenting with valvular pulmonary stenosis[1].

Pulmonic valve stenosis is characterized by varying degrees of fibrosis, thickening, and commissural fusion of the valve, resulting in restricted blood flow[5].

Symptomatic patients with moderate or severe pulmonary valve stenosis often present with exertional dyspnea or fatigue, depending on the degree of obstruction and the heart's compensatory capacity[5], as observed in our case.

In most cases, interruption of the IVC remains asymptomatic due to the well-developed azygos and hemiazygos continuation[6]. However, despite these compensatory pathways, reduced drainage from the lower extremities can lead to elevated venous pressures and stasis, thereby increasing the future risk of thrombosis[4]. Patients may also present with symptoms such as leg swelling, leg pain, lower extremity varices, abdominal pain, and, in rare instances, hematochezia[6]. In this case, none of these symptoms were observed; however, hepatomegaly accompanied by hepatojugular reflux was detected.

Sequential echocardiography can easily establish the diagnosis of interruption of the IVC[7]. Although Doppler ultrasound, magnetic resonance imaging, and computed tomography are useful diagnostic tools, they were not utilized in this case[1].

Balloon valvuloplasty via femoral venous access is the gold standard for managing isolated pulmonary stenosis[5]. However, this approach was unavailable in our case due to the interruption of the IVC. Consequently, surgical intervention was chosen. The work has been reported in line with the SCARE criteria[8].

Conclusion

It is important to know that the interruption of the IVC can occur in isolation or as part of a syndrome. Therefore, further investigations should be completed for each patient, and more research should be conducted on the associations of IVC interruption with pulmonary valve stenosis.

Ethical approval

Not applicable.

Consent

Written informed consent was obtained from the patient's parents/legal guardian for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Sources of funding

Not applicable.

Author's contribution

T.R., M.A.N., N.B., contributed to drafting, reviewing, editing, and approving the final manuscript; S.H., A.M.H.H.J., contributed to

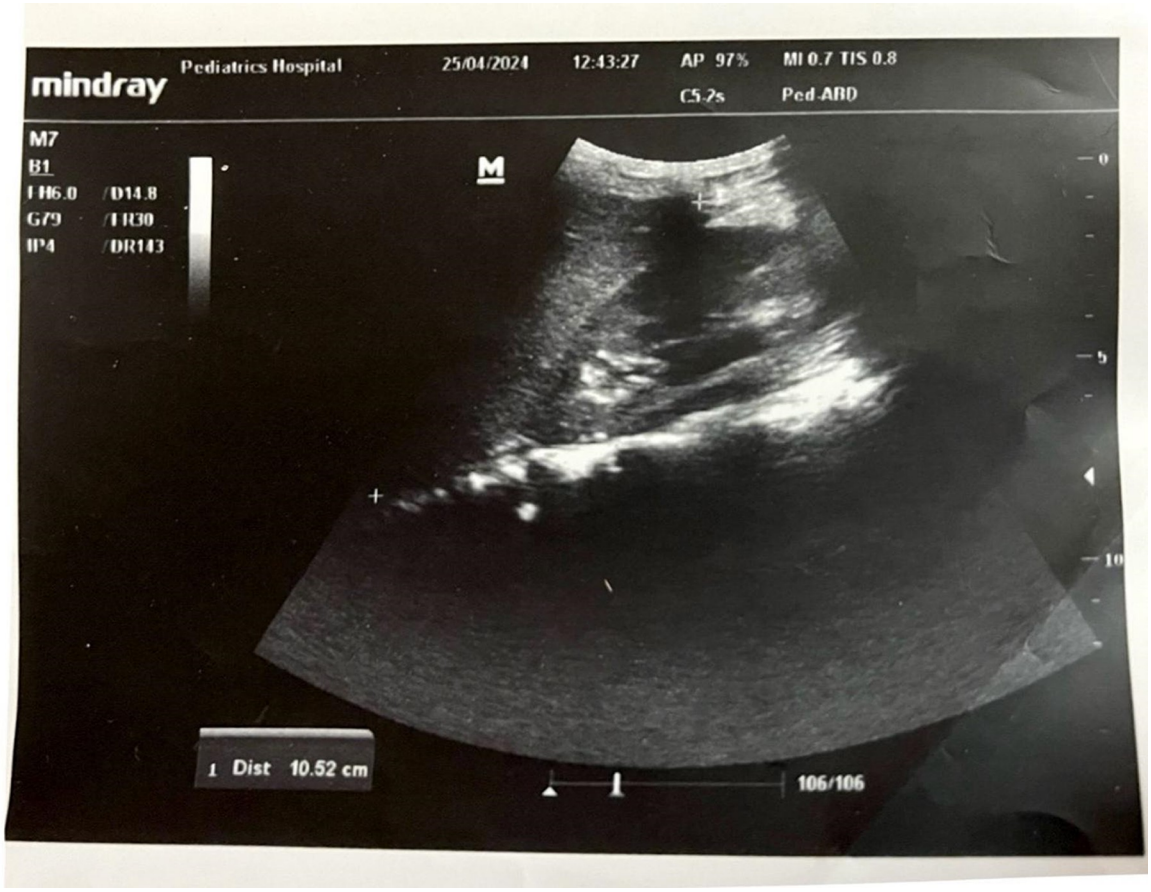

Figure 3. Abdominal ultrasound reveals the spleen to be normal and in its usual anatomical position.

drafting, editing, and approving the final manuscript; D.M.M.A., contributed to drafting, reviewing, editing, corresponding, and approving the final manuscript; S.S., contributed to reviewing, supervising, and approved the final manuscript; D.A., contributed to drafting, editing and approved the final manuscript.

Conflicts of interest disclosure

All the authors declare to have no conflicts of interest relevant to this study.

Research registration unique identifying number (UIN)

Not applicable.

Guarantor

Sultaneh Haddad.

Provenance and peer review

Not commissioned, externally peer-reviewed.

Data availability statement

Not applicable.

References

[1] Bartram U, Fischer G, Kramer HH. Congenitally interrupted inferior vena cava without other features of the heterotaxy syndrome: report of five cases and characterization of a rare entity. Pediatric and Dev Pathol 2008;11:266–73.
[2] Rajeesh G, Vaidyanathan B. Prenatal diagnosis of isolated interrupted inferior vena cava with azygos continuation to superior vena cava. Ann Pediatr Cardiol 2014;7:49–51.
[3] Obinata H, Nishibe S, Ishihara Y. Atypical perioperative management for duodenal obstruction in an infant with heterotaxy syndrome: a case report. JA Clin Rep 2018;4:16.
[4] Mansour MM, Hussein O, Saad B, *et al*. A rare case of pulmonary embolism in a patient with interrupted inferior vena cava and polysplenia. Cureus 2022;14:e22323.
[5] Heaton J, Horenstein MS, Kyriakopoulos C. Pulmonary Stenosis. In: StatPearls. StatPearls Publishing; 2024.
[6] Masino F, Muscatella G, Montatore M, *et al*. A remarkable case report of an interrupted inferior vena cava with hemiazygos and transhepatic continuation. Acta Biomed 2023;94:e2023238.
[7] Chen SJ, Wu MH, Wang JK. Clinical implications of congenital interruption of inferior vena cava. J Formos Med Assoc 2022;121:1938–44.
[8] Sohrabi C, Mathew G, Maria N, *et al*. The SCARE 2023 guideline: updating consensus surgical case report (SCARE) guidelines. Int J Surg Lond Engl 2023;109:1136.