Radiology Case Reports 20 (2025) 69–74

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

Polysplenia syndrome in adulthood: A case report of incidental discovery ☆

*Jihane El Houssni**, *Sanae Jellal*, *Ismail Neftah*, *Fariss Dehayni*, *Siham El Haddad*, *Nazik Allali*, *Latifa Chat*

Radiology Department, Pediatric Teaching Hospital, Mohammed V University, Rabat, Morocco

ARTICLE INFO

Article history:
Received 6 September 2024
Accepted 21 September 2024

Keywords:
Polysplenia syndrome
Adult
CT
Case report

ABSTRACT

The Polysplenia Syndrome (PSS) is a form of heterotaxy, a rare congenital anomaly with an estimated incidence of 1 in 250,000 live births, first described by Helwig in 1929. Most patients with polysplenia syndrome die during the neonatal period due to severe associated cardiac and biliary anomalies. Nevertheless, some individuals present with moderate cardiovascular malformations or abdominal anomalies, often discovered incidentally in adulthood. PSS is categorized under ambiguous situs syndromes or heterotaxy, also known as left or bilateral isomerism. However, it remains a complex and controversial entity, lacking specific pathognomonic features but exhibiting a broad spectrum of anomalies. We report the case of a 29-year-old woman admitted for acute chest pain, associated with lightheadedness and syncope. Clinical examination revealed tachycardia and tachypnea. A thoracoabdominal CT angiography identified 3 splenules, auricular and bronchopulmonary isomerism, along with cardiovascular and digestive anomalies, suggestive of polysplenia syndrome. Symptomatic treatment was initiated, with a favorable clinical outcome and no need for specific therapeutic intervention.

Introduction

In 1929, Helwig was the first to describe heterotaxy syndrome, more specifically polysplenia. Polysplenia syndrome (PSS) is a type of situs ambiguous, characterized by left isomerism, manifesting as the presence of multiple abnormal splenic nodules and a varied array of visceral malformations of unknown etiology. The term “left isomerism” encompasses all morphological variations of organs on the right side of the midline when they adopt certain characteristics of their left-sided counterparts [1]. Although polysplenia syndrome is primarily described in pediatrics, it is occasionally discovered incidentally in adulthood. Diagnosis is often made in childhood due to its frequent association with early-detectable cardiac malformations. Common malformations include the presence of multiple spleens, cardiac anomalies, and vascular malformations, notably involving the inferior vena cava with azygos

☆ Competing Interests: The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

* Corresponding author.
E-mail address: elhoussnijihane@gmail.com (J. El Houssni).

https://doi.org/10.1016/j.radcr.2024.09.118

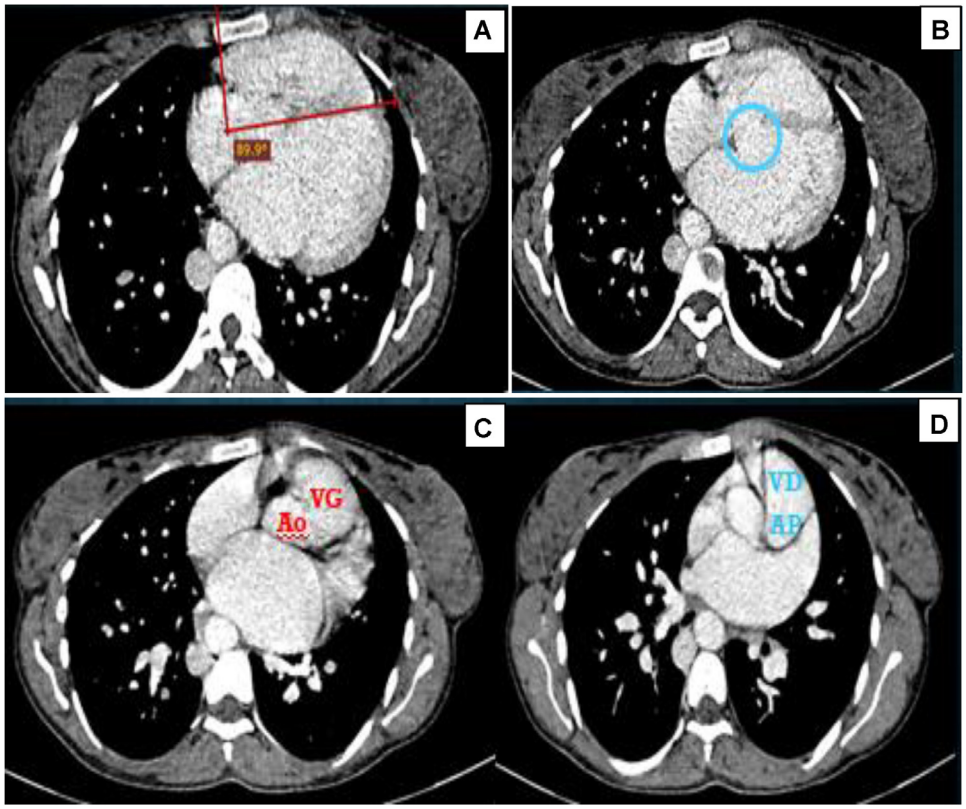

Fig. 1 – The injected thoracic CT scan of our patient revealed extreme levocardia, indicated by a deviation of the interventricular septum axis to the left of more than 65° (A), and an interventricular septal defect (B). Additionally, there was atrioventricular and ventriculoarterial concordance, indicating a normal arrangement of the major vessels (C, D).

or hemiazygos continuation [2]. In most cases, cardiac anomalies associated with PSS are moderate and nonlethal, in contrast to right atrial isomerism, which is characterized by asplenia and severe cardiac malformations. The anomalies most frequently associated with polysplenia include atrioventricular canal defects, septal malformations, anomalies of the inferior vena cava, bilateral bilobation or trilobation of the lungs, median liver position, right-sided stomach, and various degrees of intestinal malrotation [3]. Imaging advancements in ultrasound and computed tomography (CT) have significantly improved the characterization of these anomalies, aiding in the diagnostic and therapeutic management of patients [4].

Observation

A 29-year-old female patient with no significant medical or surgical history was admitted to the emergency department with acute chest pain, associated with episodes of lightheadedness and syncope. Clinical examination revealed normal blood pressure without asymmetry, tachycardia at 120 beats per minute, and tachypnea. The dextro test was normal, the ECG showed no abnormalities, and laboratory tests, including a complete blood count (CBC) and electrolyte panel, were within normal limits.

A contrast-enhanced chest CT scan revealed cardiomegaly, an interventricular septal defect, extreme levocardia, normal ventricular disposition in a D-loop configuration, and normal auriculoventricular and ventriculo-arterial concordance, indicating a normal arrangement of major vessels (Fig. 1). Observations included a bilobated right lung with hypartial bronchi, a right fissure symmetrical to the large left fissure, and dilatation of the azygos vein draining into the superior vena cava (SVC) (Fig. 2). Additionally, there was noted dilation of the hepatic veins draining into the right atrium.

In response to these radiological findings, a thoraco-abdominal angiogram was performed, which showed dilation of the left atrium and pulmonary veins, as well as dilation of the ascending aorta and the proximal descending aorta. The aortic arch displayed a Gothic configuration with an acute angle at its apex. In the abdominal region, a large azygos vein was observed replacing the inferior vena cava (IVC), with an azygos continuation of the IVC and absence of the retrohepatic portion of the IVC (Fig. 3). The liver was enlarged and positioned in the midline, the pancreas was short with agenesis of the corpo-caudal segments, and the stomach was located in the right hepatorenal space. The patient also had 3 ipsilateral splenules and a preduodenal portal trunk (Fig. 4). Based on these findings, the diagnosis for this patient is heterotaxy syndrome, specifically polysplenia syndrome.

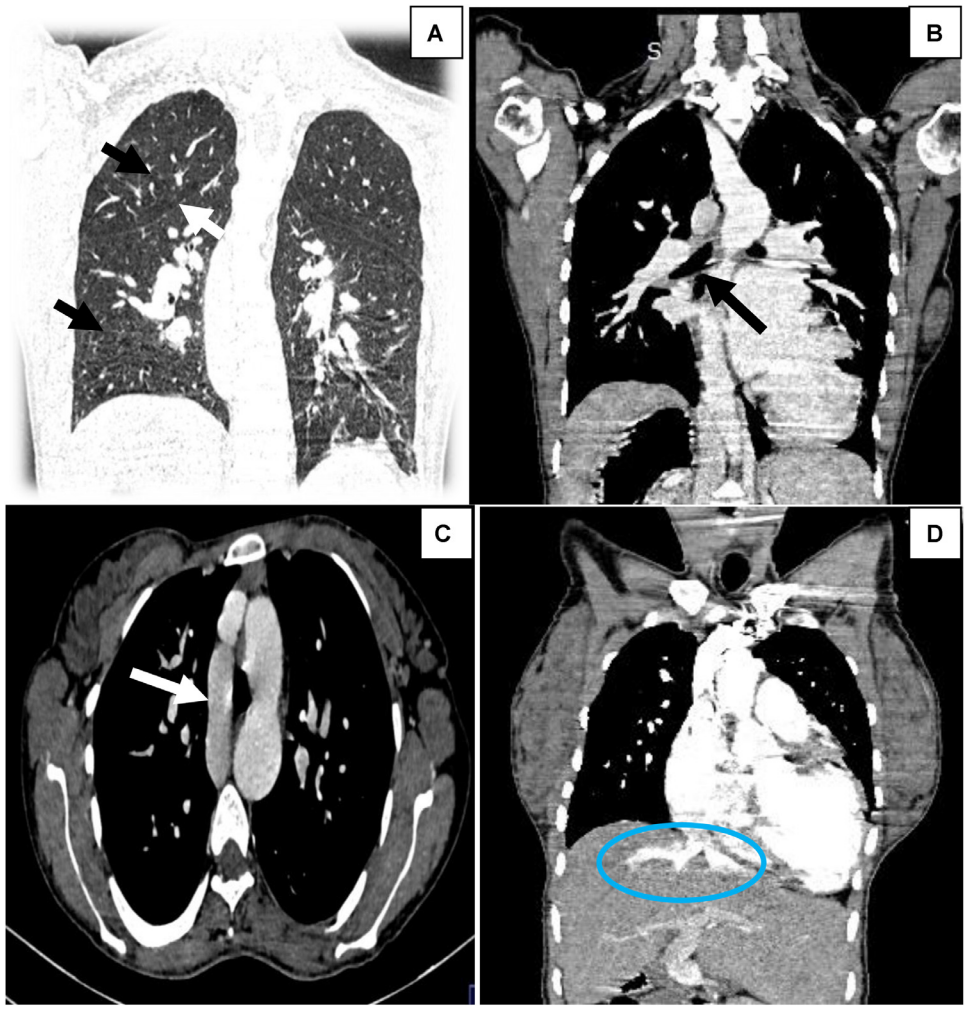

Fig. 2 – The injected thoracic CT scan showed a bilobed right lung (black arrow "A"), a right fissure symmetrical to the large left fissure (white arrow "A"), and hypartial bronchial roots (B). Additionally, there was dilation of the azygos vein draining into the superior vena cava (SVC) (C), as well as dilation of the hepatic veins draining into the right atrium (RA) (D).

Discussion

Polysplenia syndrome is a rare condition with an incidence of 1 in 250,000 live births [2]. The syndrome slightly affects more females than males [2]. It is typically associated with severe cardiovascular anomalies, leading to a 75% mortality rate by age 5. However, approximately 5%-10% of patients with polysplenia (PS) are asymptomatic and do not present with serious cardiac abnormalities [2], which may explain why polysplenia syndrome (PSS) is often diagnosed incidentally in adults, as most affected individuals show no symptoms [5]. This was the case for our patient.

The pathophysiology and molecular genetics of polysplenia syndrome are still not well understood [1]. Various genes involved in the TGF-β pathway, such as the **CFC1** gene, which encodes a component of the multi-subunit receptor for **NODAL**, have been implicated. The **CFC1** mutation has been identified in 80% of cases of left isomerism, while the **NODAL** mutation is more commonly associated with right isomerism [3]. This mutation is particularly relevant due to its association with a broad range of phenotypic changes, including polysplenia, complex cardiac anomalies, left isomerism of the lungs, bilateral superior vena cava, midline liver, right-sided stomach, and varying degrees of intestinal malrotation [3].

Imaging plays a crucial role in diagnosing polysplenia syndrome. Helical multi-detector CT angiography, with its excellent spatial and contrast resolution, is highly useful in diagnosing and detecting the various anomalies associated with polysplenia syndrome [5]. Three-dimensional volume reconstructions also allow for better analysis of vascular anomalies, which can be very helpful for surgeons if surgical intervention is considered [5].

Polysplenia syndrome is a complex condition characterized by a broad spectrum of anomalies. PSS is defined by the presence of multiple spleens, typically ranging from 2 to 6, in the absence of a history of splenectomy [5]. Some cases of polysplenia only present with a single spleen and common heterotaxy features while still being classified as

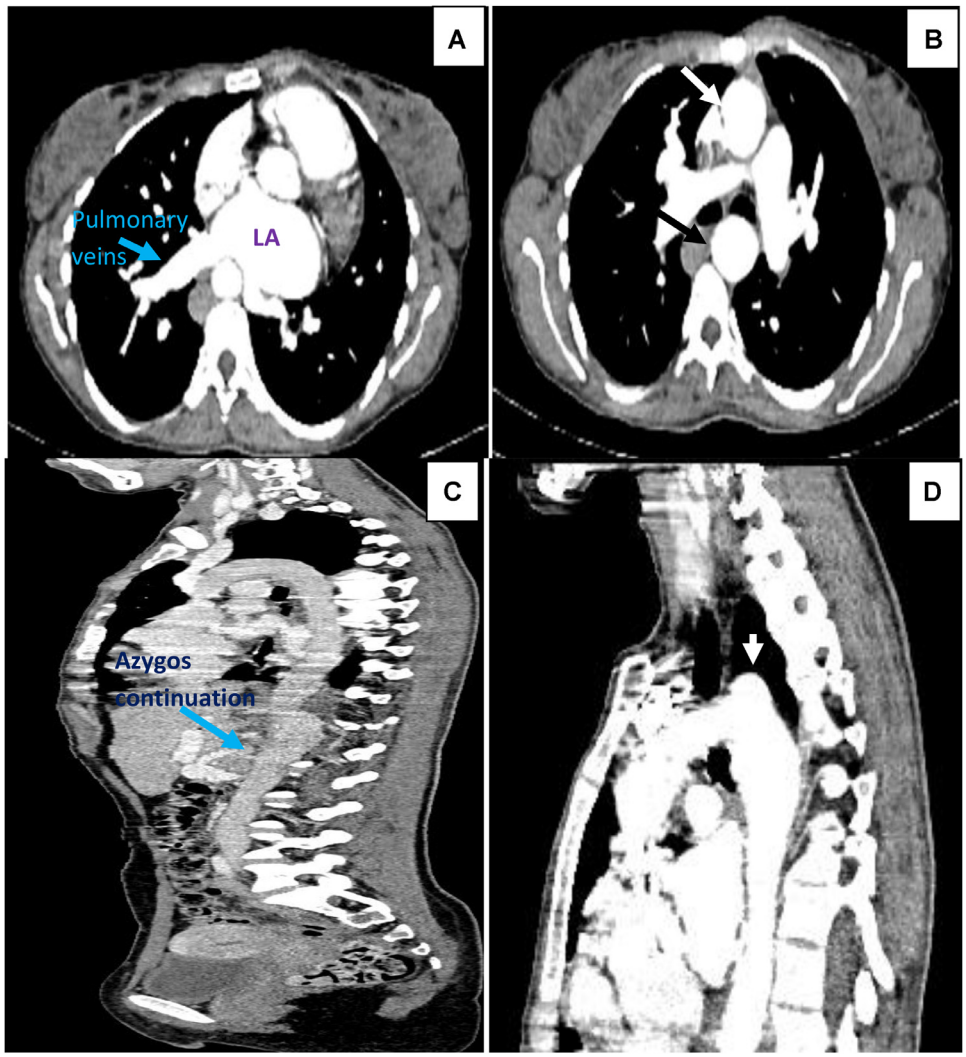

Fig. 3 – The thoraco-abdominal CTA revealed dilation of the left atrium (LA) and pulmonary veins (A), along with enlargement of the ascending aorta (White arrow "B") and the proximal portion of the descending aorta (black arrow "B"). A large azygos vein was found to replace the inferior vena cava (IVC), with azygos continuation and absence of the retrohepatic segment of the IVC (C). The aortic arch, presenting a gothic-type configuration with a sharp angle at its peak (D), requires special attention, as it may exacerbate arterial hypertension, thereby increasing the risk of perioperative hypertension.

polysplenia [2]. It is often associated with multiple congenital variations affecting the heart, solid organs, the abdominal digestive tract, or major vessels [5]. Our patient presented with 3 ipsilateral splenules, auricular and bronchopulmonary isomerism, alongside cardiovascular and digestive anomalies, suggestive of polysplenia syndrome.

Heterotaxy syndromes are known to be associated with an increased incidence of complex cardiac anomalies. In polysplenia syndrome, cardiac anomalies are generally less frequent than in asplenia. Cardiovascular abnormalities may include atrial septal defect, ventricular septal defect, bilateral superior vena cava, right aortic arch, partial anomalous pulmonary venous return, transposition of the great arteries, pulmonary valve stenosis, and subaortic stenosis [6]. In our patient, cardiovascular and pulmonary anomalies included ventricular septal defect, dilation of the azygos vein draining into the superior vena cava, and dilation of the hepatic veins draining into the right atrium.

Inferior vena cava interruption with azygos continuation is the second most common anomaly after multiple spleens in polysplenia syndrome [2,6]. This anomaly results from a failure in the right subcardinal-hepatic anastomosis, leading to atrophy of the subcardinal vein (adrenal segment) and continuation of the infrarenal IVC as an azygos vein. The suprahepatic segment of the IVC is generally present and drains separately into the right atrium [6].

Since the dorsal pancreatic bud and spleen develop in the dorsal mesogastrium, anomalies in these 2 organs may occur in patients with polysplenia syndrome. Studies frequently report cases of a short or truncated pancreas, where only the head of the pancreas, sometimes with a small part of the body, is present. Clinically, this can lead to an increased incidence

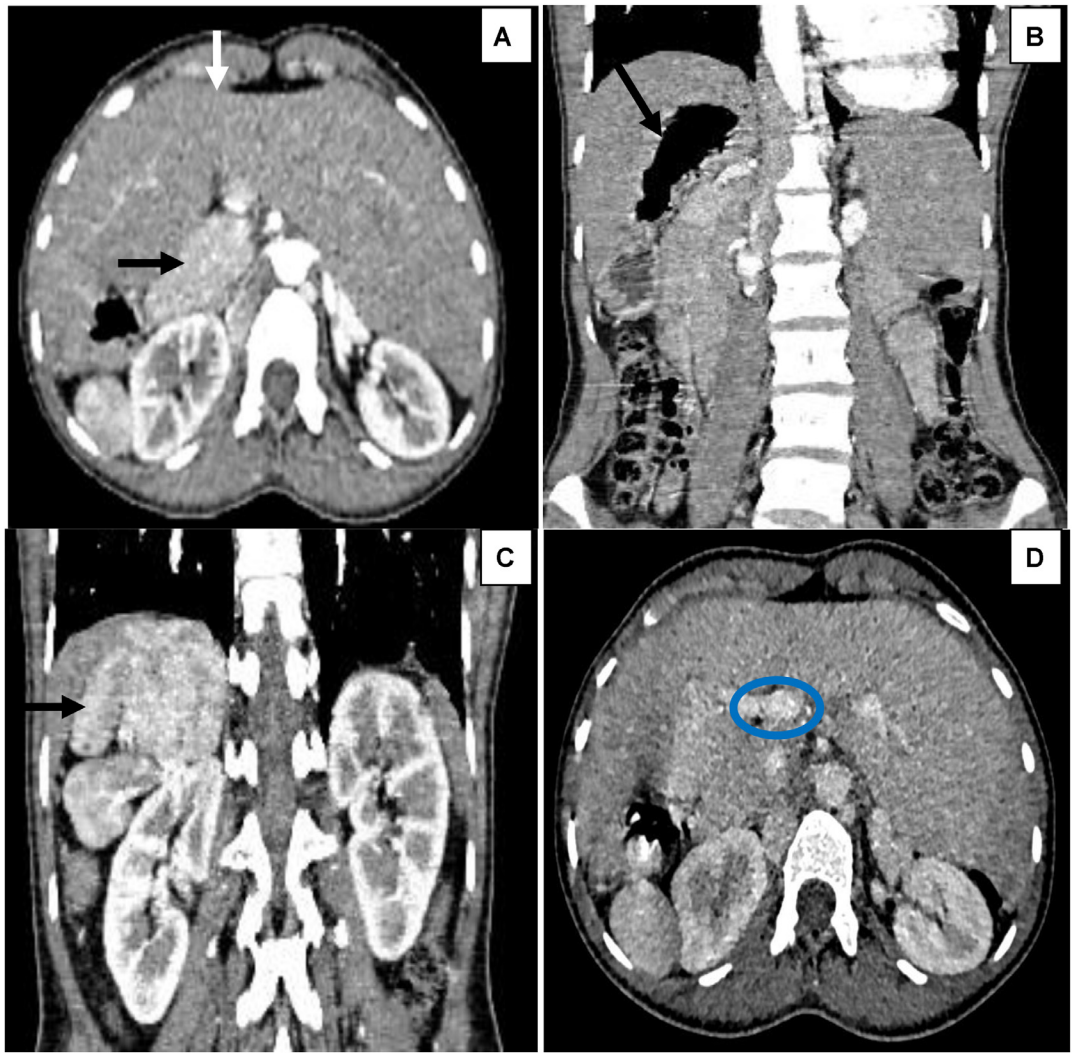

Fig. 4 – Abdominal CTA revealed an enlarged liver in a midline position (white arrow "A"). The pancreas was short, with agenesis of the corpo-caudal segments (black arrow "A"). The stomach was located in the right hepato-renal space (B), along with the presence of 3 ipsilateral splenules (C). Additionally, a preduodenal portal vein was identified (D).

of pancreatitis and diabetes mellitus [6]. Other described pancreatic anomalies include short pancreas, annular pancreas, pancreatic divisum, and pancreatic malrotation [2,7].

The presence of a midline liver, bilobate bilateral lungs, hypartial bronchi, and bilateral pulmonary veins in PSS is noted, although these are not systematically associated with polysplenia [6]. Another anomaly associated with polysplenia syndrome is the preduodenal portal vein. This venous anomaly is common in the syndrome, passing ventrally relative to the duodenum and head of the pancreas, and appears as a round structure anterior to the head of the pancreas on CT and MRI images [1]. Generally, this anomaly has no significant clinical impact unless encountered during biliary or hepatic surgical procedures [6].

Intestinal malrotation is observed in 60.4% of polysplenia cases [2]. Radiological detection of these malrotations is crucial, even in the absence of symptoms, as predicting patients at risk of volvulus can be challenging. Surgical correction is generally recommended for all patients with malrotation [6].

Other anomalies reported in PSS include poly-lobed gallbladder, biliary atresia, and genitourinary malformations such as renal agenesis, hypoplastic kidneys, and duplication of the collecting systems, which were fortunately absent in this case [1]. In our patient, none of these anomalies were observed.

Management of polysplenia syndrome depends on the patient's age. In childhood, surgical intervention is often necessary to address cardiac anomalies, which generally have a poor prognosis. In adulthood, management is symptomatic, with particular attention to anomalies such as the preduodenal portal vein, intestinal malrotation, or vascular anomalies to prevent complications during surgical interventions [2,8]. Our patient received only symptomatic treatment.

Conclusion

In conclusion, polysplenia syndrome is a complex condition characterized by a broad spectrum of anomalies, with the

most common being multiple spleens and inferior vena cava interruption with azygos continuation. This rare hereditary syndrome often presents in childhood, although its discovery in adulthood is typically incidental. In adulthood, polysplenia syndrome generally does not require specific treatment.

Patient consent

Written informed consent was obtained from the patient for their anonymized information to be published in this article

REFERENCES

[1] Plata-Muñoz José, Hernández-Ramírez D, Javier Anthón F, Podgaetz E, Avila-Flores F, Chan C. Polysplenia syndrome in the adult patient. Case report with review of the literature. Ann Hepatol 2004;3(3):114–17. doi:10.1016/s1665.

[2] Malki MC, Outznit M, Mechhor S, Bouibaouen B, Nkurunziza L, Bacha HE, et al. Polysplenia syndrome in adulthood: a case report. Pan Afr Med J 2022;41:67. doi:10.11604/pamj.2022.41.67.29014.

[3] Mohor CI, Fleaca SR, Oprinca Muja A, Oprinca GC, Roman MD, Chicea R, et al. A rare case of polysplenia syndrome associated with severe cardiac malformations and congenital alveolar dysplasia in a one-month-old infant: a complete macroscopic and histopathologic study. J Cardiovasc Dev Dis 2022;9(5):135. doi:10.3390/jcdd9050135.

[4] Mountassir ME, Borahma M, Benelbarhdadi I, Lagdali N, Hosni A, Ajana FZ. Polysplenia syndrome revealed in adulthood by pancreatic and vascular malformations: a case report. Pan Afr Med J 2022;43:77. doi:10.11604/pamj.2022.43.77.31496.

[5] Liu J, Yang K, Wang J. Polysplenia syndrome with a rare variation between the common hepatic artery and the superior mesenteric artery in adults. Arch Med Sci 2020;16(5):1263–6. doi:10.5114/aoms.2020.97972.

[6] El Mortaji H, Elatiqi K, El Hammaoui H, Alj S. Polysplenia syndrome with situs ambiguous, common mesentery, and IVC interruption discovered incidentally in an adult. Radiol Case Rep 2019;14(9):1072–5. doi:10.1016/j.radcr.2019.05.032.

[7] Abdullah RA, Alharbi MA, Alnutaifi FM, Faqih AI, Alduraibi KM, Alharbi AS. Polysplenia syndrome complicated by splenic infarction: a report of a rare case. Cureus 2023;15(5):e38589. doi:10.7759/cureus.38589.

[8] Tawfik AM, Batouty NM, Zaky MM, Eladalany MA, Elmokadem AH. Polysplenia syndrome: a review of the relationship with viscero-atrial situs and the spectrum of extra-cardiac anomalies. Surg Radiol Anat 2013;35(8):647–53. doi:10.1007/s00276-013-1100-x.