


Polysplenia Syndrome With Persistent Left Superior Vena Cava: Case Report and Review of the Literature

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El Houss Salma, MD¹ , Amsguine Najwa, MD¹, Tantaoui Mehdi, MD¹, Allali Nazik, PhD¹, Chat Latifa, PhD¹, and El Haddad Siham, PhD¹

Abstract

Polysplenia syndrome is an uncommon condition associating several splenic nodules (sometimes polylobed spleen and cases of normal spleen have been described) with a number of malformations that appear between the fourth and sixth week of embryonic development. Although it has been suggested that genetic, teratogenic, and embryogenic factors may be at fault, the exact etiology remains unclear. Clinically, it is generally asymptomatic or mildly symptomatic. The authors report the case of an 11-months-old infant from a poorly monitored pregnancy. He was admitted to the emergency room for respiratory discomfort in a context of apyrexia. A thoraco-abdominal CT scanner revealed a polysplenia syndrome.

Keywords

polysplenia, ambiguous situs, heterotaxy, mesocardia, agenesis of the inferior vena cava, CT

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Case Report

An 11-month-old boy, from a poorly monitored pregnancy, got operated on for a common atrioventricular canal. After a 6 months, he was brought to the emergency room with respiratory discomfort, no fever and no other history according to the mother and the referring physician. At admission, the infant was conscious and responsive but hypotonic, with an oxygen saturation level of 89%. The patient was started on oxygen therapy and a chest radiograph was performed. It showed some alveolar opacities of both lung fields, which prompted us to realize a thoraco-abdominal CT scan. On the mediastinal window, we discovered an situs ambiguous with a mesocardia, a medial liver, a spleen replaced by several right splenic nodules and a right stomach (Figure 1); with left isomerism: both atria were of left morphology and a hyperarterial bronchi (Figure 2). In addition to that, we found associated anomalies of venous return such as the azygos continuation of the inferior vena cava, which was agenetic in its retro-hepatic part, with hepatic veins flowing directly into the patient's right atrium (Figures 3 and 4). Moreover, the superior vena cava was located on the left (Figure 5) receiving both the azygos vein (Figures 6 and 7)

and the innominate trunk on the left. While the ascending and descending aortas were in place (Figure 5). Furthermore, no abnormalities of the pulmonary arteries or the common mesentery were noted. On the parenchymal window, both lungs were bilobed with mosaic perfusion pattern of the two lung fields (Figure 8).

Discussion

Heterotaxy syndrome¹ is an anomaly of distribution of thoracic and abdominal organs. In this entity, the usual left-right distribution of these organs (situs solitus) does not correspond entirely to a situs inversus (complete mirror image). It is thus called situs ambiguus.

Polysplenia syndrome² is a rare form of heterotaxy syndrome. Also known as left isomerism, it is characterized by a number of spleens greater than or equal to

¹Mohammed V University, Rabat, Morocco

Corresponding Author:

El Houss Salma, Department of Radiology Mother and Child, Ibn Sina Hospital, Mohammed V University, Rabat, 12000, Morocco.
Email: alma.elhous94@gmail.com





Figure 1. 11-month-old male infant with polysplenia syndrome.

Findings: Abdominal CT scan in injected axial section showing a median liver (a) with right splenic nodules (b) and a right stomach.
Technique: Philips, 23 mL IV ultravist 300, kV 110, mA 93.



Figure 3. 11-month-old male infant with polysplenia syndrome operated for commonatrioventricular canal.

Findings: Chest CT in injected axial section showing a medial liver with suprahepatic vein (c).
Technique: Philips, 23 mL IV ultravist 300, kV 110, mA 93.

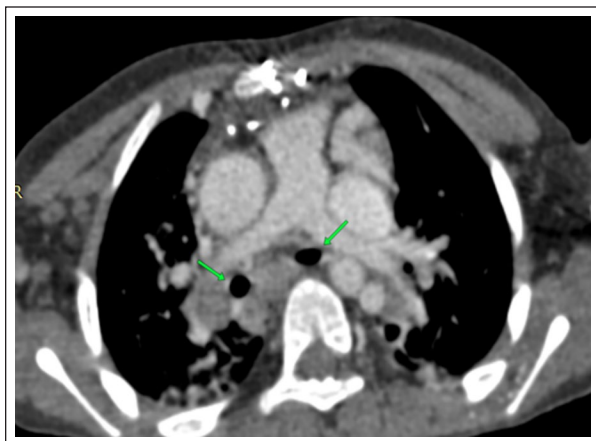


Figure 2. 11-month-old male infant with polysplenia syndrome operated for common atrioventricular canal.

Findings: Abdominal CT scan in injected axial section showing a bilateral hyperarterial bronchi (green arrows).
Technique: Philips, 23 mL IV ultravist 300, kV 110, mA 93.

two with identical volume, and a lateralization anomaly in the form of situs ambiguus; although situs inversus might exceptionally be encountered. Some forms of polysplenia syndromes can have a single normal or polylobed splenic gland. Moreover, considering the embryology of the splenic tissue that develops in the posterior mesogastrium, the spleen and or the splenic nodules are always located on the same side of the stomach along the greater curvature.

Other anomalies include an agenesis of the supra-renal IVC with a continuous azygos system and a direct drainage of the hepatic veins into the right atrium. Some authors

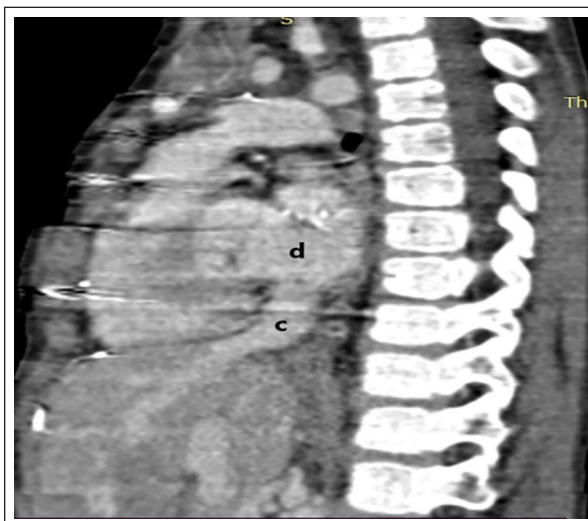


Figure 4. 11-month-old male infant with polysplenia syndrome operated for common atrioventricular canal.

Findings: Sagittal section reconstruction of an injected thoracic CT scan showing agenesis of the inferior vena cava with a suprahepatic vein (c) draining directly into the right atrium (d).
Technique: Philips, 23 mL IV ultravist 300, kV 110, mA 93.

also described associations with other cardiac, pulmonary, vascular and digestive malformations.²

Etiology and Demographics (Table 1)

It is predominant in females with an estimated incidence of 1 per 250 000 live births. And it can be diagnosed during adulthood as well as childhood.³



Figure 5. 11-month-old male infant with polysplenia syndrome operated for common atrioventricular canal. Findings: Thoracic CT scan in injected axial section showing ascending (e) and descending (f) aorta in place with venous return anomaly such as azygos substitution (g) of the inferior vena cava with the superior vena cava which is left (h). Technique: Philips, 23 mL IV ultravist 300, kV 110, mA 93.



Figure 7. 11-month-old male infant with polysplenia syndrome operated for common atrioventricular canal. Findings: Thoracic CT scan in injected axial section showing a mesocardia with the descending aorta (f), the azygos vein on the left (g) and agenesis of the inferior vena cava. Technique: Philips, 23 mL IV ultravist 300, kV 110, mA 93.



Figure 6. 11-month-old male infant with polysplenia syndrome operated for common atrioventricular canal. Findings: Thoracic CT in injected axial section objectifying the azygos vein (g) located on the left which flows into the superior vena cava which is left (h). Technique: Philips, 23 mL IV ultravist 300, kV 110, mA 93.



Figure 8. 11-month-old male infant with polysplenia syndrome operated for common atrioventricular canal. Findings: Chest CT scan with parenchymal window showing a mosaic lung with several frosted glass areas in relation to a perfusion disorder. Technique: Philips, 23 mL IV ultravist 300, kV 110, mA 93.

Peoples et al⁴ described the first case of polysplenic syndrome in 1781. After performing a series of 146 autopsies, they assessed the prevalence of the most frequently found abnormalities. 58% of patients had bilateral bilobed lungs with left-type bronchial segmentation, 47% had bilateral superior vena cava, more than 60%

had cardiac abnormalities, and 56% had gastrointestinal positional abnormalities.

Although no clear etiology has been identified yet, some clues tend toward embryogenic, genetic and teratogenic causes.⁵ Furthermore, mutations have been identified in the genes of patients with heterotaxia.

Table 1. Summary Table of Polysplenia Syndrome.

Etiology	<ul style="list-style-type: none"> • Not yet fully understood • Factors embryogenic, genetic, and teratogenic have been suggested
Incidence	<ul style="list-style-type: none"> • 1 per 250 000 live births has polysplenia syndrome
Gender predilection	<ul style="list-style-type: none"> • Female gender
Age predilection	<ul style="list-style-type: none"> • Childhood and adulthood
Risk factors	<ul style="list-style-type: none"> • Factors embryogenic, genetic, and teratogenic • Mutations have been identified in the genes of patients with heterotaxia.
Treatment	<ul style="list-style-type: none"> • The surgical treatment is indicated according to the type of associated cardiac and vascular malformations also according to the age and the clinical tolerance
Prognosis	<ul style="list-style-type: none"> • Generally good prognosis but can sometimes depend on essentially cardiac malformations
Findings on imaging	<ul style="list-style-type: none"> • Situs ambiguous or rarely situs inversus • Polysplenia sometimes a polylobed or normal spleen • Left isomerism • Agenesis of the inferior vena cava with continuation of azygos type
	Often
	<ul style="list-style-type: none"> • Bilobed right lung • Common atrioventricular canal • Complete common mesentery and other malformations . . .

Clinical and Imaging Findings (Table 1)

The malformations generally found in polysplenia syndrome appear between the fourth and sixth week of embryonic life²:

Cardiovascular anomalies are represented by a defect of the inter-ventricular or inter-auricular septum, a common atrioventricular canal, a double outlet of the left ventricle, a common atrium, interposition of the portal vein in preduodenal, transposition of the large vessels, sometimes a double superior vena cava and rarely persistence of the left superior vena cava without individualization of the right inferior vena cava (which was the case in our patient).²

Digestive anomalies include a complete common mesentery, an annular pancreas, a microcolon, gallbladder agenesis (50%) and/or biliary atresia, which in some cases requires a liver transplant.

Most pulmonary anomalies consist of bilateral bilobed lungs with a left type segmentation since it is a left isomerism malformation. Trilobed lungs, having a right type segmentation, are generally associated with asplenia, another kind of heterotaxy syndrome. This asplenia syndrome or Ivemark syndrome is characterized by a right isomerism and is a differential diagnosis of the polysplenia syndrome.

Thanks to the viable cardiac malformations, polysplenia syndrome can remain asymptomatic and diagnosis is

usually fortuitous in adulthood. Whereas in asplenia syndrome, these cardiac malformations are lethal.⁶

Prenatal ultrasound can help in prenatal diagnosis, revealing essentially lateralization anomalies.

Imaging techniques, including thoraco-abdominal CT scan with contrast, are essential for the minutiose assessment of the malformations as well as for the pre-operative strategy.

Treatment and Prognosis (Table 1)

Surgical treatment is adapted to the type of cardiac and vascular malformations and to age and clinical tolerance.

In case of atrioventricular canal; as the case was for our patient; surgical management will be adjusted to the age (2 and 6 months) as well as to the valvular leaks and the pulmonary vascular resistances).

Prognosis depends on the morbidity and mortality of the associated malformations, especially congenital heart disease. As a matter of fact, prognosis of these cardiopathies is generally good thus the overall favorable outcome seen in polysplenia syndrome patients unlike asplenia syndrome.

Differential Diagnoses (Table 2)

Asplenia syndrome is a type of heterotaxy syndrome (situs ambiguous) which is characterized by asplenia, right isomerism, trilobate left lung, superior vena cava duplication, complex congenital heart disease, and gastro-intestinal anomalies.

Table 2. Differential Diagnosis Table for Polysplenia Syndrome.

Differential diagnosis	Clinical	CT
Polysplenia syndrome	<ul style="list-style-type: none"> Asymptomatic or minimally symptomatic Intestinal malrotation with midgut volvulus may also be a presenting feature. Symptoms of postoperative complications not included 	<ul style="list-style-type: none"> Situs ambiguous or rarely situs inversus Polysplenia sometimes a polylobed or normal spleen Left isomerism Agenesis of the inferior vena cava with continuation of azygos type <p>Often</p> <ul style="list-style-type: none"> Bilobed right lung Common atrioventricular canal Complete common mesentery and other malformations . . .
Asplenia syndrome	<ul style="list-style-type: none"> Cyanotic congenital heart disease is the main presentation 	<ul style="list-style-type: none"> Heterotaxia (situs ambiguous) Asplenia Right isomerism Trilobed left lung Duplication of the superior vena cava Complex congenital heart disease like single ventricle Gastrointestinal anomalies . . .

Abbreviations: CT, computed tomography; IVC: inferior vena cava.

Conclusion

Polysplenia syndrome is a rare polymalformative condition associating vascular, cardiac, pulmonary, and visceral malformations. CT imaging is a must in the minutiose assessment of the malformations spectrum, thus making it easier to make an accurate prognosis and eventually plan for surgical management when necessary.

Teaching Point

The importance of this article lies in presenting and describing a unique form of heterotaxy syndrome: polysplenia syndrome with an unusual left inferior vena cava. As well as showing the importance of imaging in the diagnosis and the evaluation of associated malformations for a clearer prognosis, a more precise preoperative workup and early surgical treatment of the cardio-vascular abnormalities.

Authors Contributions

El Houss Salma: Preparation and creation of the published work. Amsiguine Najwa: Preparation and creation of the published work. Tantaoui Mehdi: Preparation and creation of the published work. Chat Latifa: Supervising published work. Allali Nazik: Supervising published work. El Haddad Siham: Supervising published work.

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Consent

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ORCID iD

El Houss Salma  <https://orcid.org/0000-0003-0725-054X>

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Questions

- ✓ Question 1: Polysplenia syndrome is a rare congenital pathology: (Which of the following answers is true)
 - 1-Not included in the heterotaxy syndrome
 - 2-The inferior vena cava is always present
 - 3-Can present with a single normal spleen (applies)
 - 4-Is a right isomerism
 - 5-Always associated with a bilobed right lung
 - Explanation for question 1:
 - 3: Polysplenia syndrome may be associated with the presence of a single normal spleen. [some polysplenic syndromes have been described with a single normal or polylobed splenic gland].
- ✓ Question 2: The polysplenia syndrome: (Which of the following answers is true)
 - 1-Frequently affects the female sex (applies)
 - 2-Has an incidence of 1 birth/100 000
 - 3-Affects only the child
 - 4-Associated with several types of malformations other than cardiac (applies)
 - 5-The associated heart defects are not viable
 - Explanation for question 2:
 - 1: The female sex is the most frequently affected in the syndrome Polysplenia syndrome [It is a syndrome predominantly in female patients].
 - 4: Polysplenia syndrome is associated with several types of cardiac, vascular, and digestive malformations [associated with other cardiac, pulmonary, vascular, and digestive malformations]
- ✓ Question 3: Asplenia syndrome: (Which of the following answers is true)
 - 1-Is a syndrome of heterotaxy (applies)
 - 2-Also called ivermak syndrome (applies)
 - 3-Asplenia is constantly present (applies)
 - 4-Is a left isomerism
 - 5-Differential diagnosis of polysplenia syndrome (applies)
 - Explanation for question 3:
 - 1: The asplenia syndrome includes an ambiguous situs and is therefore part of the heterotaxy syndrome [Another type of heterotaxy syndrome which is the asplenia syndrome].
- 2: Asplenia syndrome is also called ivermak syndrome [the asplenia syndrome (Ivemark syndrome)]
- 3: Asplenia is part of the definition of asplenia syndrome [An asplenia entering within the framework of another type of heterotaxy syndrome which is the asplenia syndrome]
- 5: The main differential diagnosis of asplenia syndrome is the polysplenia syndrome because the similarity of most anomalies found [And is a differential diagnosis of the polysplenic syndrome].
- ✓ Question 4: Concerning the anomalies of laterali-sation (Which of the following answers is true)
 - 1-The spleen is not systematically located on the side of the stomach
 - 2-Can be diagnosed antenatally (applies)
 - 3-Are part of the polysplenia syndrome (applies)
 - 4-Correspond to the situs ambiguus alone
 - 5-Situs inversus type of lateralization anomalies rarely found in polysplenia syndrome (applies)
 - Explanation for question 4:
 - 2: Prenatal ultrasound allows to detect organ positional anomalies and therefore allows to diagnose lateralisation anomalies [Prenatal diagnosis by prenatal ultrasound can reveal lateralization anomalies].
 - 3: Polysplenia syndrome is a syndrome of heterotaxy, so the lateralisation anomalies are part of it [The polysplenia syndrome is a type of heterotaxis syndrome also known as left isomerism which is a rare congenital condition with a number of spleens greater than or equal to 2 (of identical volume), a lateralization anomaly]
 - 5: The situs ambiguus is the main lateralisation anomaly associated with the polysplenia syndrome because it is part of the heterotaxy syndrome. [a lateralization anomaly (mainly situs ambiguus as it is a heterotaxis syndrome, situs inversus is part of the lateralization anomalies exceptionally encountered in this syndrome and a rarely described association)]

- ✓ Question 5: polysplenia syndrome (Which of the following answers is true)
 - 1-Is of very well established etiology
 - 2-Is always symptomatic
 - 3-The malformations appear between the fourth and sixth week of embryonic's life. (applies)
 - 4-Has a good prognosis in general (applies)
 - 5-In case of associated common atrio-ventricular canal surgical treatment can be considered even after 6 months of life
 - Explanation for question 5:
 - 3: Malformations associated with polysplenia syndrome are present between

the fourth and sixth week of embryonic's li [The malformations generally found in the polysplenia syndrome appear between the fourth and sixth week of embryonic life].

- 4: The cardiac anomalies associated with polysplenia syndrome are generally viable and not complex which provides this syndrome a good prognosis [generally this syndrome has a good prognosis due to the fact that associated cardiopathies have a good overall prognosis].