

Atypical presentation of scimitar syndrome with severe hepatomegaly: a case report

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Background

Scimitar syndrome is a rare congenital disease characterized by partial or total anomalous pulmonary venous return from the right lung into the systemic venous system, and accounts for 0.5–2% of all congenital heart disease. Severe forms of the disease are diagnosed in childhood. However, because of the benign form of the syndrome in adults, many are asymptomatic, or present only mild symptoms including exertional dyspnoea, arrhythmias, and respiratory infections. We report an atypical presentation with hepatomegaly.

Case summary

A 24-year-old woman was evaluated for abdominal discomfort. Physical examination revealed a remarkable hepatomegaly. Chest X-ray revealed dextroversion, enlargement of the right cavities, and a curvilinear opacity known as ‘scimitar sign’. A transthoracic echocardiography demonstrated right ventricular dilation and a venous collector draining into right suprahepatic vein, which was severely dilated, with large hepatomegaly. Scimitar syndrome was confirmed by magnetic resonance imaging (MRI). Therefore, the patient underwent surgery, redirecting the pulmonary venous return to left atrium. Three months later, the patient remained asymptomatic and both the hepatomegaly and the right chamber volumes normalized.

Discussion

Abdominal discomfort, as in our clinical case, is a highly atypical presentation of scimitar syndrome. It is important for physicians to be aware that diagnostic suspicion can be established from a chest X-ray, on which the scimitar sign can be distinguished in many cases. The diagnosis must be confirmed with other imaging modalities, such as echocardiography, MRI, or computed tomography. Corrective surgery may relieve the symptoms related to liver congestion at follow-up.

Keywords

Case report • Scimitar syndrome • Anomalous pulmonary venous return • Hepatomegaly • Cardiac magnetic resonance

Learning points

- Scimitar syndrome includes a wide spectrum of clinical manifestations that vary from an incidental finding in asymptomatic adults to atypical symptoms such as hepatomegaly and abdominal discomfort.
- The diagnosis can be suspected by characteristic features on chest X-ray; however, it is critical to systematically perform an echocardiogram in every patient.
- Corrective surgery with baffle repair or scimitar vein reimplantation into the left atrium may relieve abdominal symptoms and resolve hepatomegaly and right chamber enlargement at follow-up.

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Introduction

Scimitar syndrome is a rare congenital disease characterized by partial or total anomalous pulmonary venous return (APVR) from the right lung into the systemic venous system, causing a shunt from left to right.¹ It is associated with other malformations including hypoplasia of the right lung and cardiac dextroversion. The syndrome can present early in the neonatal period or later in life. When it presents in childhood, patients often exhibit severe symptoms, such as heart failure, pulmonary hypertension, recurrent respiratory infections, or cyanosis. However, adults with scimitar syndrome, who often exhibit a minor form, can remain asymptomatic or mildly symptomatic, usually with mild exertional dyspnoea.^{2,3}

We report an unusual case with atypical presentation, in which the right pulmonary veins drained into the right suprahepatic vein causing abdominal discomfort with severe hepatomegaly, which was resolved by surgery.

Timeline

- Before admission: Abdominal discomfort and exertional dyspnoea
- October 2017
 - Electrocardiogram revealed an incomplete right bundle branch block with ST segment changes in leads V1–V3
 - Chest X-ray suggested dextroversion and enlargement of the right chambers with the 'scimitar sign'
 - Echocardiography demonstrated a venous collector from right pulmonary veins to right suprahepatic vein, with dilation of the suprahepatic vein and right chambers
- November 2017
 - Magnetic resonance imaging (MRI) confirmed anomalous right pulmonary venous drainage with hepatomegaly and severe dilation of suprahepatic veins. A direct suprahepatic venous drainage was observed in the right atrium and the right ventricle was dilated with normal systolic function
- January 2018
 - Right heart catheterization demonstrated a significant shunt (Q_p/Q_s ratio of 2/1) and ruled out the presence of pulmonary hypertension
- August 2018
 - Surgery was performed using an intra-atrial patch to baffle the flow of the scimitar vein into the left atrium through a newly created atrial septal defect
- October 2018
 - The patient remained asymptomatic after surgery and follow-up MRI revealed normalization of the right chamber volumes and significant reduction in hepatomegaly and calibre of the suprahepatic veins

Case presentation

A 24-year-old Hispanic woman with no relevant medical history was evaluated for epigastric fullness and abdominal discomfort accompanied by mild exertional dyspnoea. Her vital signs were stable, although physical examination revealed significant, non-painful, pulsatile hepatomegaly with no other remarkable findings.

Electrocardiogram revealed an incomplete right bundle branch block, with ST segment changes in leads V1–V3. Chest X-ray suggested dextroversion and enlargement of the right cavities. There was also a curvilinear opacity that extended from the right pulmonary hilum, bordering the cardiac silhouette to the homolateral cardiophrenic angle, which is known as the 'scimitar sign' (Figure 1). Results of liver function tests were within the normal range.

A transthoracic echocardiogram demonstrated right pulmonary veins draining directly into the right suprahepatic vein, which was severely dilated. There was also atypical hepatic venous drainage, with independent suprahepatic veins into the right atrium (Figure 2 and Video 1). This was associated with mild pulmonary hypertension [estimated mean pulmonary artery pressure (mPAP) 35 mmHg] and mild right ventricle (RV) dilation (RV end-diastolic basal diameter, 45 mm) with normal systolic function (Video 2). The presence of atrial septal defect (ASD) was ruled out by transoesophageal echocardiography.

Cardiac magnetic resonance imaging (MRI) confirmed RV dilation [end-diastolic volume (EDV), 130 mL/m²; end-systolic volume (ESV), 42 mL/m²] and identified the venous collector of the entire right lung ending in right suprahepatic vein, with shunt to middle suprahepatic vein and an increased pulmonary to systemic blood flow ratio (Q_p/Q_s 1:9). Both veins were severely dilated with large hepatomegaly but no systemic venous collaterals were detected. In addition, the concomitant presence of an intraparenchymal venous malformation

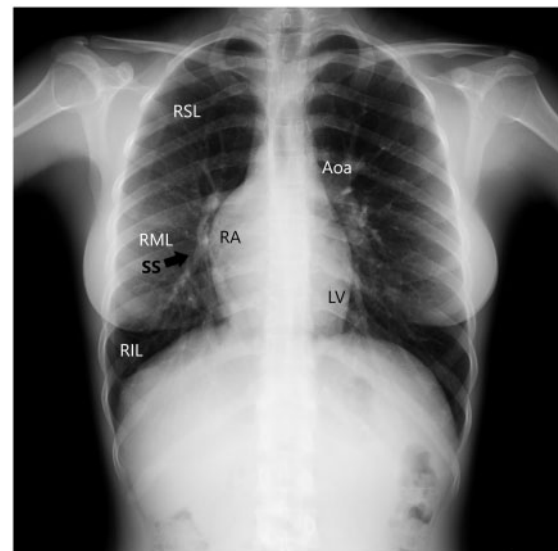


Figure 1 Chest X-ray revealing the scimitar sign (black arrow), dextroversion, and right cavity enlargement. AOA, aortic arch; LV, left ventricle; RA, right atrium; RIL, right inferior lobe; RML, right middle lobe; RSL, right superior lobe; SS, scimitar sign.

with abundant tortuous and dilated veins was observed. It also revealed right lung hypoplasia, with the absence of upper lobar bronchus and mediastinal shift towards the right hemithorax (Figure 3). Angiography was also performed, which confirmed the presence of a venous collector that provided drainage of the three pulmonary lobes, but no systemic collaterals were identified (Video 3).

Subsequently, right heart catheterization was performed, in which the presence of pulmonary hypertension was ruled out, with an mPAP of 21 mmHg and normal pulmonary vascular resistance (2 Wood Units), and confirming the presence of a significant Qp:Qs ratio of 2. Accordingly, surgery was indicated because the patient exhibited symptomatic anomalous pulmonary venous drainage accompanied by overloading of right cavities and remarkable hepatomegaly. She underwent surgery using an intra-atrial patch to baffle the flow from the anomalous pulmonary venous system into the left atrium through the right atrium, creating an ASD. The postoperative course was uneventful, and the patient was discharged on Day 6 in good clinical condition.

Three months after surgery, the patient remained asymptomatic and follow-up MRI revealed normalization of the right chamber volumes (EDV, 73 mL/m²; ESV, 27 mL/m²), as well as a significant reduction in hepatomegaly and calibre of the suprahepatic veins (Figure 4).

Discussion

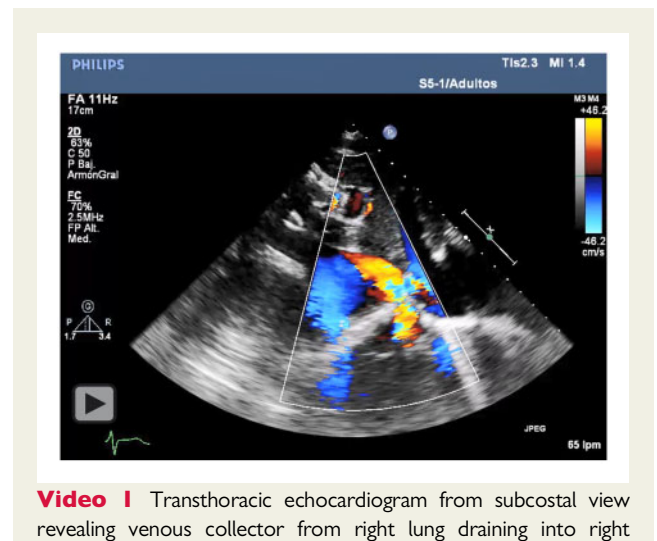
Scimitar syndrome is a complex form of APVR, an anatomy first described by Cooper and Chassin in 1836, and accounts for 3–5% of APVRs.¹ However, the true incidence may be higher because, in adulthood, many patients remain asymptomatic.

This syndrome rarely presents as an isolated abnormality. Most frequently, it is associated with hypoplasia of right lung and dextrocardia,² as observed in our case. It is also usually associated with hypoplasia of the right pulmonary artery, collaterals to the right lung from the descending aorta, and sinus venosus type ASD, all of which were absent in our patient.

When diagnosed in childhood, this entity is more likely to be associated with severe symptoms, such as respiratory failure, cyanosis, or respiratory infections, thus conferring a poorer prognosis. If the patient exhibits aortopulmonary collaterals to the sequestered lung, heart failure and pulmonary hypertension are common. However, when diagnosed in adults, it is usually associated with milder symptoms such as progressive dyspnoea, and may even be detected as an incidental finding on imaging tests.³ In our case, the presentation was highly atypical; more specifically, abdominal discomfort caused by severe volume overload in the suprahepatic veins.

Several congenital heart diseases may present with hepatomegaly, including tetralogy of Fallot, Ebstein's anomaly, transposition of the great vessels, or those with single ventricle physiology. However, hepatomegaly is rare in scimitar syndrome, especially in the absence of pulmonary hypertension or right ventricular dysfunction.

The diagnosis is established by demonstrating venous return from the right lung to the systemic venous system, usually to the hepatic



Video 1 Transthoracic echocardiogram from subcostal view revealing venous collector from right lung draining into right suprahepatic vein.

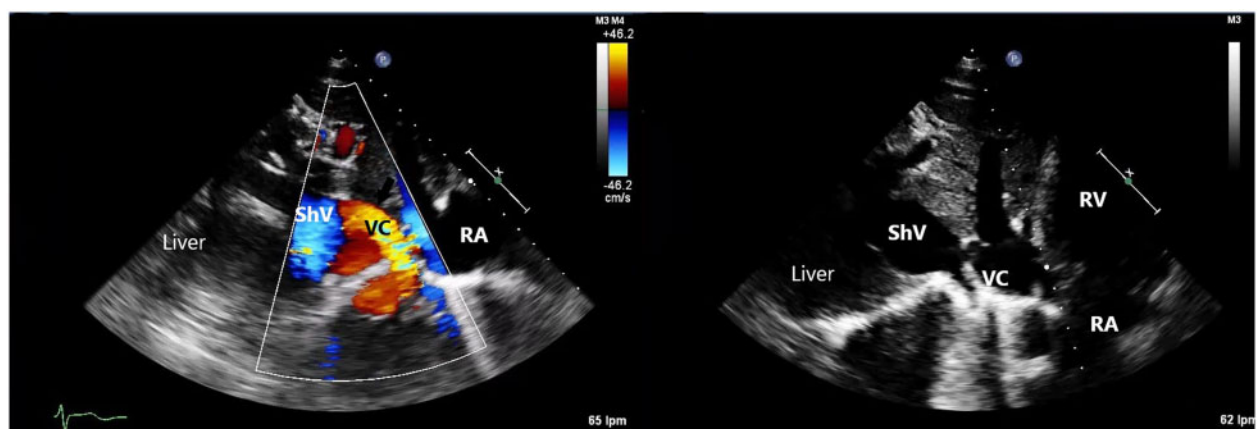


Figure 2 Transthoracic echocardiogram from subcostal view revealing venous collector (marked with black arrow) from right lung draining into suprahepatic veins, which are severely dilated. RA, right atrium; RV, right ventricle; ShV, suprahepatic vein; VC, venous collector.

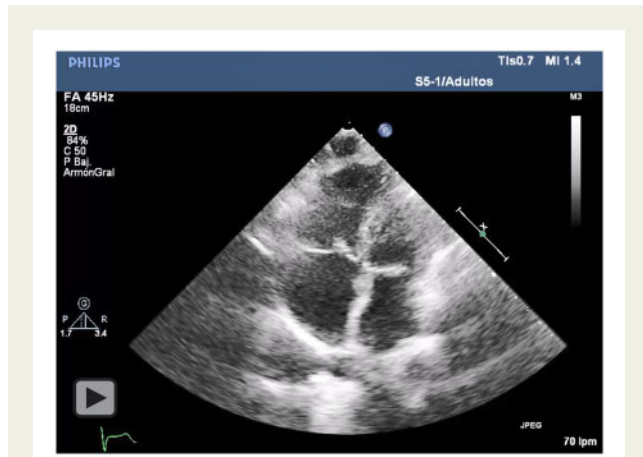
portion of the inferior vena cava. However, it can also drain into the suprahepatic veins, as in our case, and even into the portal vein, azygos, or directly into the right atrium. In our patient, the collector provided drainage of the entire lung; however, in one-third of cases, it can drain only the lower portion of the right lung, with a normorelated upper pulmonary vein.

Multiple imaging modalities, including echocardiography, MRI, and/or computed tomography (CT), can be used.⁴⁻⁶ However, it is

important to be aware that diagnostic suspicion can also be established from a simple chest X-ray, on which the scimitar sign can be distinguished in many cases.

The most available modality and, therefore, the one that should be performed systematically as the first-line choice is echocardiography; however, it also requires considerable experience to assess drainage of the pulmonary veins. CT angiography is currently the modality of choice for non-invasive assessment of vascular pathologies of the chest, and also enables evaluation of the lung parenchyma. However, MRI is also a good alternative that avoids exposure to ionizing radiation. It provides high-quality thoracic vascular images and is the modality of choice for evaluating volumes. In our case, the diagnosis was suspected on chest X-ray and, because it was a young woman, we used echocardiogram as the initial modality, completing the study using MRI.

Indications for surgery include the presence of symptoms, ASD accompanying the scimitar vein, volume overload of right chambers, and/or the presence of stenosis of the abnormal right pulmonary venous trunk.^{7,8} Our patient not only presented with overload in the right chambers, but also exhibited volume overload at the suprahepatic level, which precipitated the hepatomegaly and, consequently, the atypical abdominal symptoms. Therefore, our patient underwent surgery and symptoms disappeared, and hepatomegaly and right ventricular volume were normalized.



Video 2 Transthoracic echocardiogram from apical view revealing right ventricular dilation with normal systolic function.

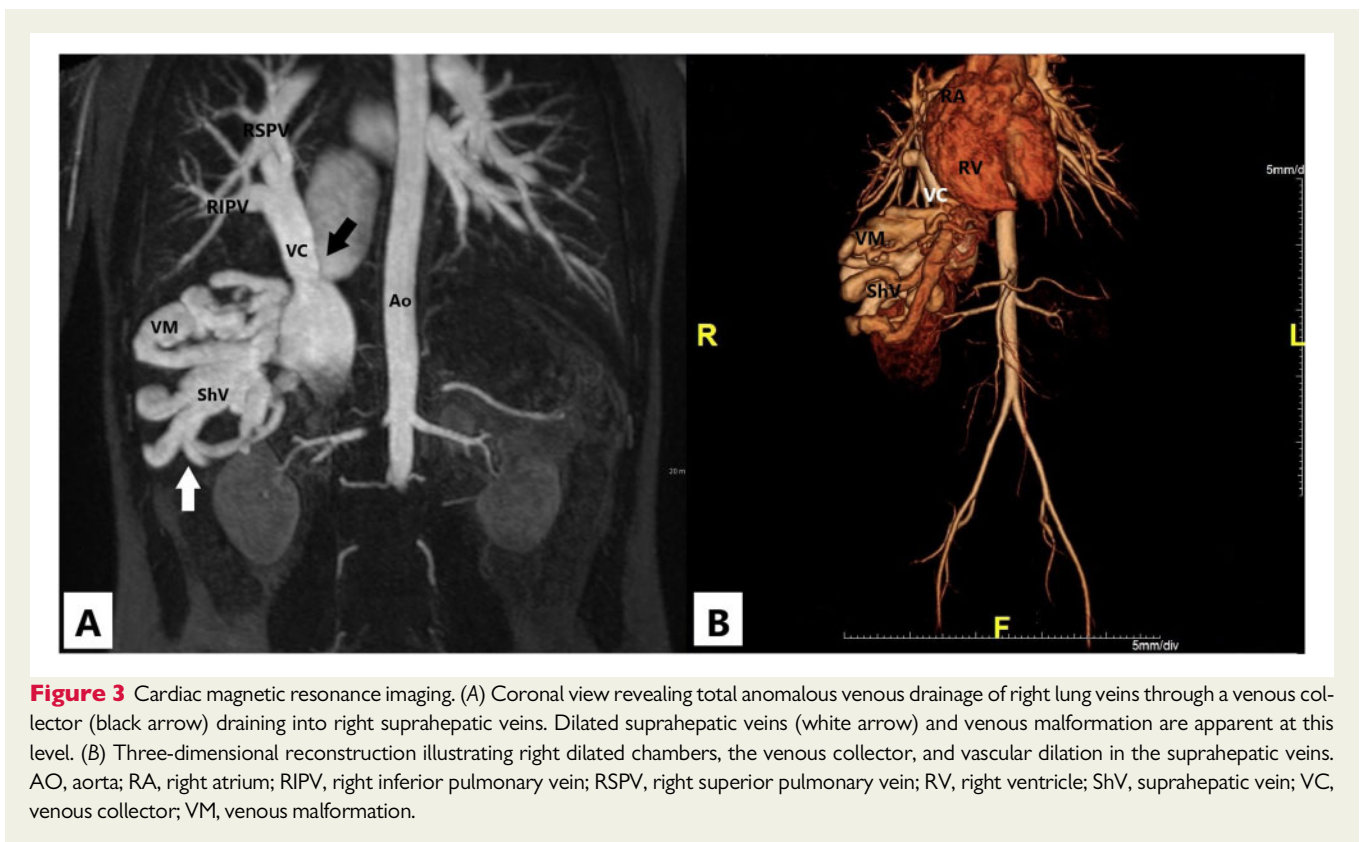
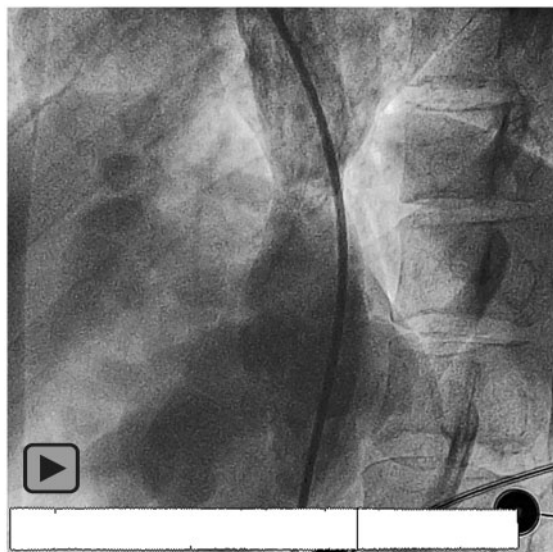


Figure 3 Cardiac magnetic resonance imaging. (A) Coronal view revealing total anomalous venous drainage of right lung veins through a venous collector (black arrow) draining into right suprahepatic veins. Dilated suprahepatic veins (white arrow) and venous malformation are apparent at this level. (B) Three-dimensional reconstruction illustrating right dilated chambers, the venous collector, and vascular dilation in the suprahepatic veins. AO, aorta; RA, right atrium; RIPV, right inferior pulmonary vein; RSPV, right superior pulmonary vein; RV, right ventricle; ShV, suprahepatic vein; VC, venous collector; VM, venous malformation.



Video 3 Angiography revealing a venous collector that provided drainage of the three pulmonary lobes, ending in right suprahepatic vein.

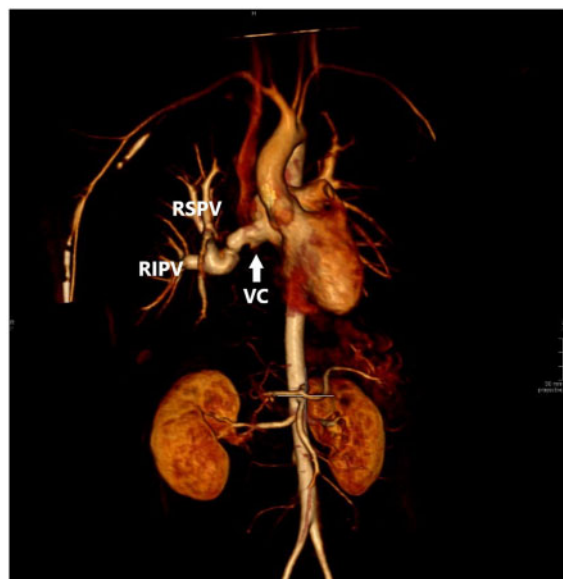


Figure 4 Cardiac magnetic resonance imaging. Three-dimensional reconstruction illustrating redirection of drainage from the right lung to the left atrium (white arrow), with normalization of right chamber volume. RIPV, right inferior pulmonary vein; RSPV, right superior pulmonary vein; VC, venous collector.

Lead author biography



Dr Alba Cruz-Galbán was born in Madrid, Spain, and studied Medicine at University Complutense of Madrid, Spain, where she was qualified in 2016. She is currently a fellow at the University Hospital of Salamanca, Spain, training to become a registered cardiologist. Her particular interests lie in the field of congenital heart disease.

Supplementary material

Supplementary material is available at *European Heart Journal - Case Reports* online.

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We sought the services of Editage for English language editing.

Slide sets: A fully edited slide set detailing this case and suitable for local presentation is available online as [Supplementary data](#).

Consent: The authors confirm that written consent for submission and publication of this case report including images and associated text has been obtained from the patient in line with COPE guidance.

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