CASE REPORT

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Scimitar syndrome associated with aberrant right subclavian artery, diaphragmatic hernia, and urinary anomalies – case report and review of the literature

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Abstract

Scimitar syndrome is a form of a partially or totally right pulmonary venous return to the inferior vena cava, which may associate variably right lung hypoplasia, right pulmonary artery hypoplasia, pulmonary sequestration together with the presence of aortopulmonary collaterals from the descending aorta towards the right lung. In many cases, there are also other cardiac anomalies associated. We present a unique association of a partially anomalous pulmonary venous return to the inferior vena cava with other vascular and thoracic anomalies: inferior sinus venosus and secundum atrial septal defect, retroesophageal right subclavian artery, obstructed accessory right bronchus, diaphragmatic hernia with ectopic liver, "S"-type thoracic scoliosis and malformations of the urinary tract (duplication of the right ureter and of the left basinet). The patient had a reimplantation of the "scimitar" vein to the left atrium and closure of the inferior sinus venosus and secundum atrial septal defect.

Keywords: scimitar syndrome, aberrant right pulmonary venous return, aberrant right subclavian artery, inferior sinus venosus defect, diaphragmatic hernia.

Introduction

Scimitar syndrome (SS) is a syndrome characterized by specific features: partial or total right anomalous pulmonary venous return to the inferior vena cava (IVC), right pulmonary artery hypoplasia, atrial septal defect (ASD), right lung hypoplasia, aorto-pulmonary collateral vessels, diaphragmatic hernia, horseshoe lung [1–4]. The presence of a diaphragmatic hernia may be a consequence of an intrauterine developmental disorder implying the involution of a lung segment. Two forms of SS are described. The infantile form has other vascular and thoracic malformations associated [3]. The adult form usually is asymptomatic and is assimilated with an incomplete form of SS of an isolated partially anomalous pulmonary venous return to the IVC without the other components.

We present the case of an unique association of partially anomalous pulmonary venous return to the IVC with other vascular and thoracic anomalies: inferior sinus venosus and secundum atrial septal defect, retroesophageal right subclavian artery, obstructed accessory right bronchus, diaphragmatic hernia with ectopic liver, "S"-type thoracic scoliosis and malformations of the urinary tract (duplication of the right ureter and of the left basinet).

Case presentation

A 9-year-old girl, diagnosed at the age of 5 with a right diaphragmatic hernia, visited our Clinic for a headache, sweating, fatigue, and paresthesia of the right limbs that began six months prior to admission in the Department of Surgery. Clinical examination on admission revealed a comfortable patient with insignificant findings at clinical examination except for mild decreased oxygen saturation (93%) and the presence of a second-degree cardiac systolic murmur with rhythmic heart sounds. Laboratory tests on blood were normal.

The chest X-ray (Figure 1A) showed enlarged pulmonary hilum, increased area of the pulmonary artery trunk, increased dimensions of the lower left arch and a circular opacity with well-defined cranial contour interpreted as being the right diaphragmatic hernia, confirmed by computed tomography (CT) scan (Figure 1B). Electrocardiogram showed rsR' pattern in V1, suggesting right ventricle overload, with prolonged QT interval (Figure 2), which disappeared subsequently.

CT scan

CT scan also revealed multiple malformations of the heart, aorta, lungs, diaphragm, liver, kidney and spine (Figure 3, A–C).

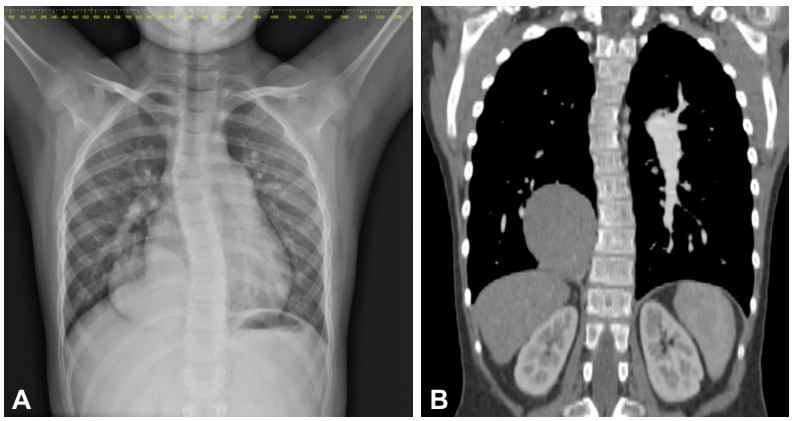

Figure 1 – *(A) Chest X-ray – scoliosis; a diaphragmatic hernia, hypervascularization of the lung, dilated pulmonary trunk, scimitar vein; (B) Computed tomography – "S"-type thoracic scoliosis, without any other alteration of the vertebrae.*

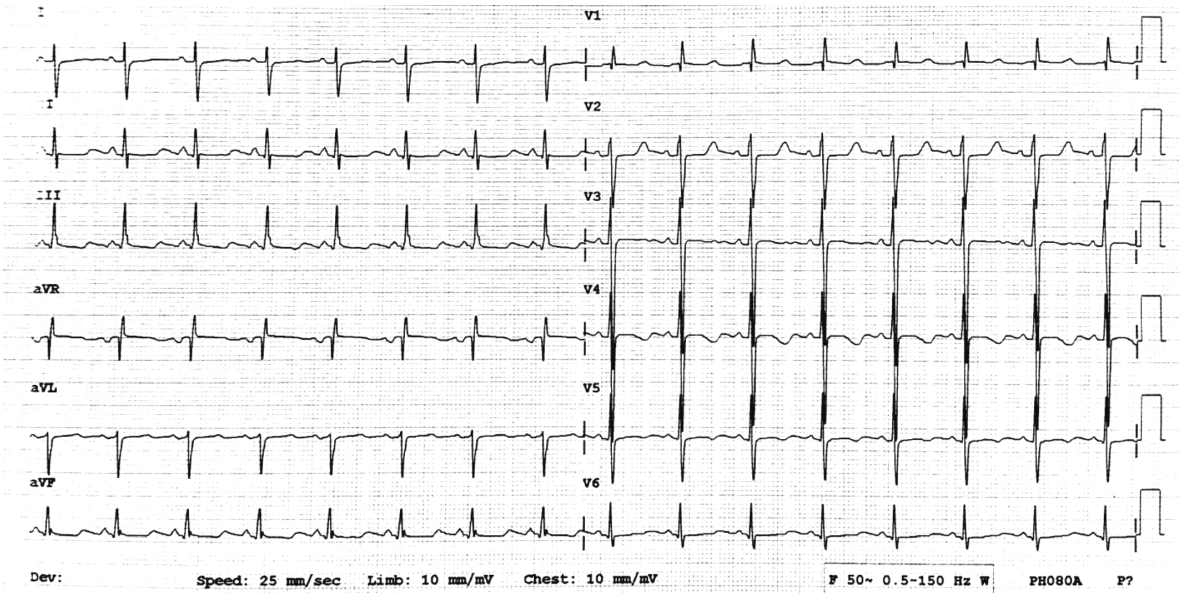

Figure 2 – *Electrocardiogram. Right axis deviation. Right ventricle overload (qR pattern in V1, transition zone in V5). Prolonged QT.*

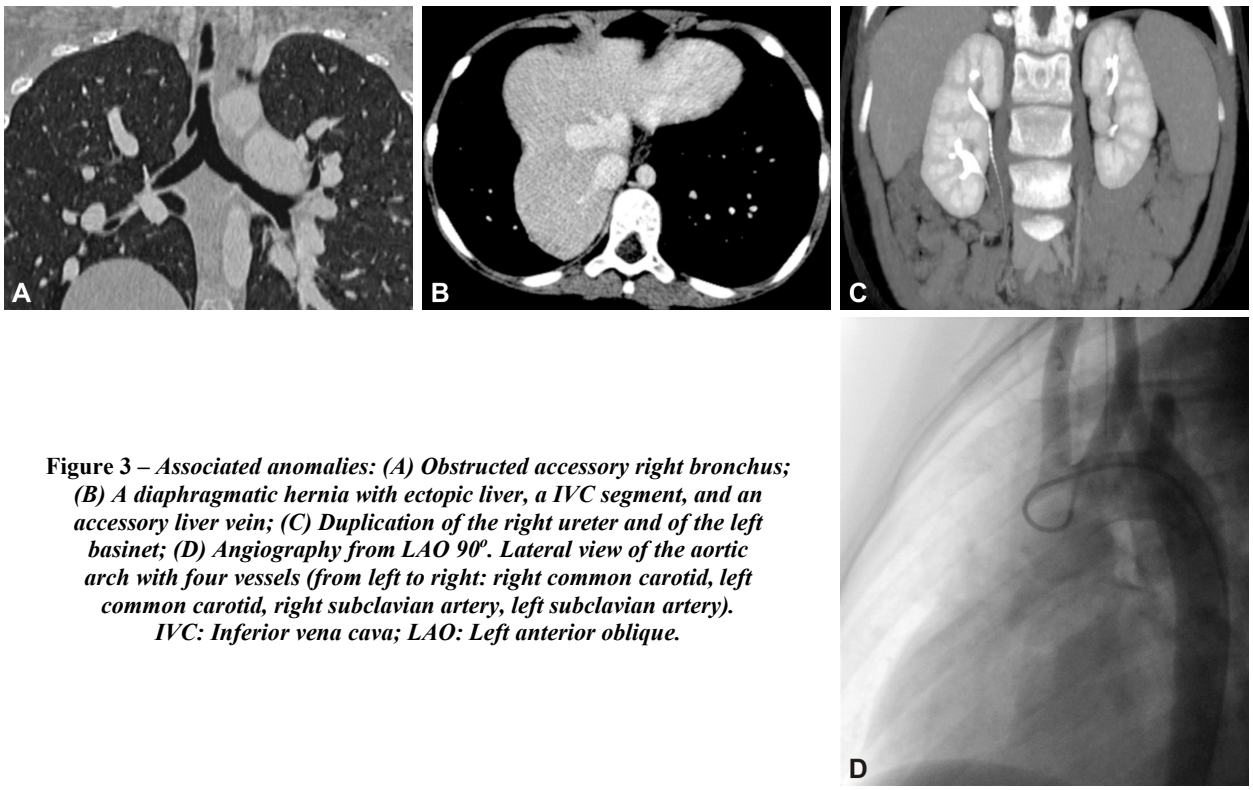

Figure 3 – *Associated anomalies: (A) Obstructed accessory right bronchus; (B) A diaphragmatic hernia with ectopic liver, a IVC segment, and an accessory liver vein; (C) Duplication of the right ureter and of the left basinet; (D) Angiography from LAO 90º. Lateral view of the aortic arch with four vessels (from left to right: right common carotid, left common carotid, right subclavian artery, left subclavian artery). IVC: Inferior vena cava; LAO: Left anterior oblique.*

A collector of the inferior and middle right pulmonary veins was described, which was attached to the confluence of the inferior vena cava to the right and left atrium – no interatrial septum was noticed (Figure 4, A and B). Besides the abnormal drainage of the right pulmonary veins, obstructed right bronchus, diaphragmatic hernia containing hepatic tissue, ASD, retroesophageal right subclavian artery, scoliosis and malformations of the ureteral tree in both kidneys with ureteral duplication on the right side and basinet bifidity were described. The right diaphragmatic hernia had a postero-medial position and contained hepatic tissue together with an accessory hepatic vein.

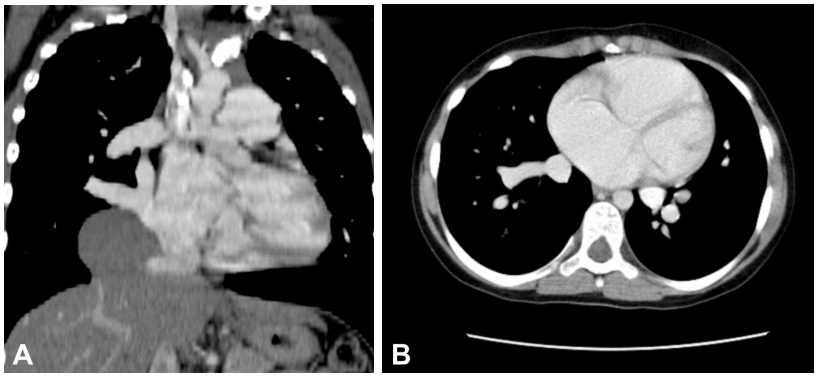

Figure 4 – ***Angio CT: (A) The middle and lower right pulmonary veins draining through a descending collector vein (scimitar vein) at the junction of the inferior vena cava to the right atrium; (B) Right heart dilatation without an evident interatrial septum. Scimitar vein directed towards the confluence of the inferior vena cava and the right atrium. CT: Computed tomography.***

Echocardiography

Echocardiography revealed a large interatrial septal defect (ASD) associated with enlarged right atrium and ventricle. The systemic veins drainage was normal. The drainage of the left pulmonary veins was into the left atrium, while the right pulmonary veins were draining at the confluence of the IVC to the right atrium. A large Eustacchio valve (EV) was visualized at the entrance of the IVC in the right atrium. The pulmonary artery trunk and branches were dilated. The pulmonary systolic arterial pressure was estimated on the base of the tricuspidian regurgitant jet at 40 mmHg. The aortic arch had a particular aspect of the right subclavian artery coming from the descending aorta and a retroesophageal route. Another cardiac pathology was excluded (Figure 5, A–D).

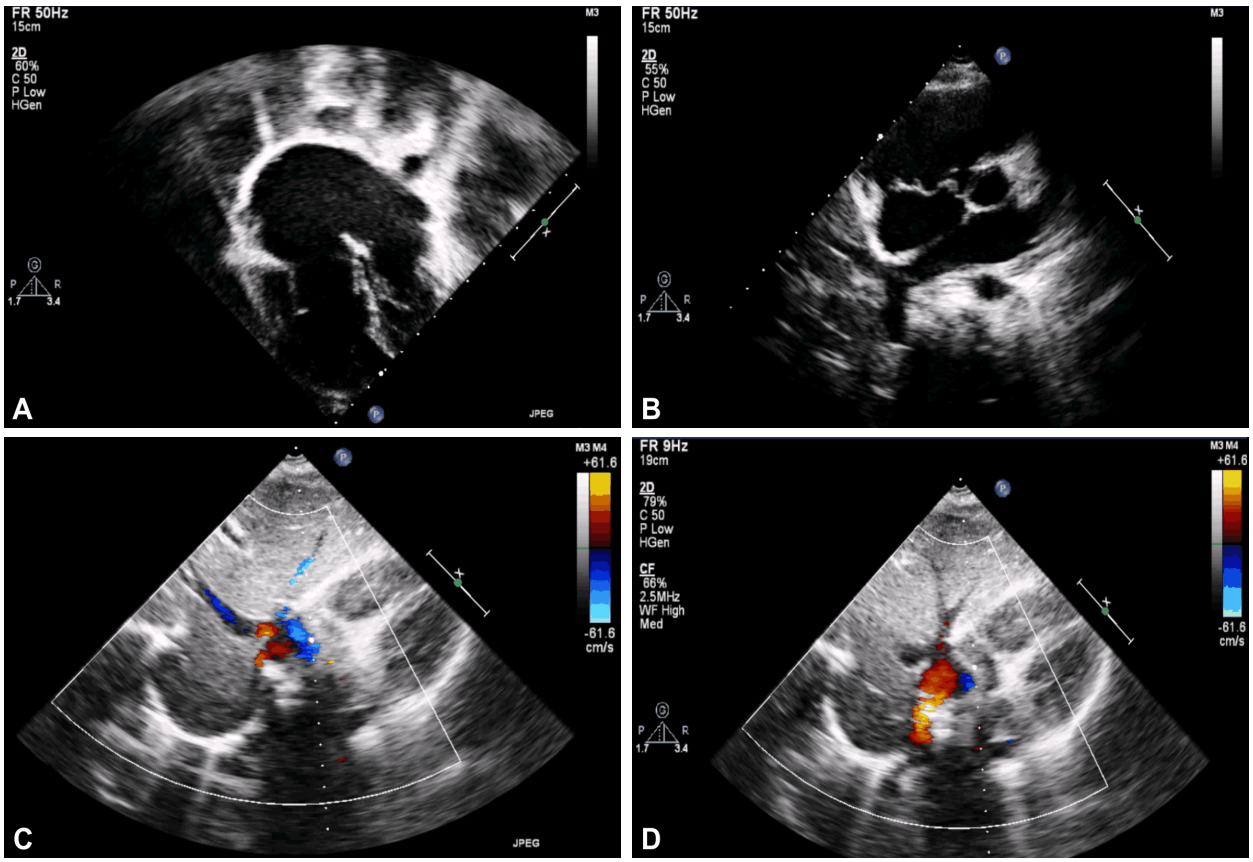

Figure 5 – ***Echocardiography: (A) Apical four chambers view, right heart dilation, large ASD; (B) Parasternal short axis – the confluence of the scimitar vein (vertical vein) to the IVC; (C) Subcostal view – suprahepatic vein draining into the IVC; (D. Subcostal view – scimitar vein draining into the IVC. ASD: Atrial septal defect; IVC: Inferior vena cava.***

Cardiac diagnostic catheterization

Further on, a cardiac diagnostic catheterization was performed to demonstrate the heart anatomy and to evaluate the pulmonary pressure and resistance in order to establish the best therapeutic approach. The angiograms showed that the upper right pulmonary vein drained directly into the right atrium, while the middle and lower right pulmonary veins were draining through a descending collector vein (scimitar vein) at the junction of the inferior vena cava to the right atrium. In addition, a large ASD as a common atrium with a bidirectional shunt, and a lusorian artery were noticed (Figure 3D).

Corrective surgery

Corrective surgery was performed with wide opening of the right pleural cavity. The right venous pulmonary collector was identified at the junction between IVC and right atrium. The venous pulmonary collector was separated from the IVC. Through a right inferiorly extended atriotomy a wide ASD, inferior sinus venous without posterior edge was identified, but with well-represented anterior and superior rims. The venous pulmonary collector was mobilized into the pericardial cavity. An incision was made on the upper wall of the pulmonary venous collector, then on the corresponding wall of the left atrium, and the two walls were sutured to each other, thus expanding the mouth of the pulmonary collector. Closure of the ASD with pericardial bovine patch and reversing the pulmonary collector to the left atrium were done. The EV was partially resected in order to increase the opening of the inferior vena cava. The diaphragmatic hernia, which had a postero-medial location, could not be approached by sternotomy. Also, the asymptomatic retroesophageal right subclavian artery was not corrected. After the surgery, the recovery was favorable.

The postprocedural echocardiography showed the ASD closing patch with a small residual shunt, the right pulmonary vein collector redirected to the left atrium with unobstructed, laminar flow, mildly accelerated flow at the junction of the inferior vena cava to the right atrium, but with normal venous velocities, good kinetics and no pericardial effusion (Figure 6, A and B). Seven days after the procedure, the patient was discharged hemodynamically stable, with good cardiac, respiratory, digestive and renal function.

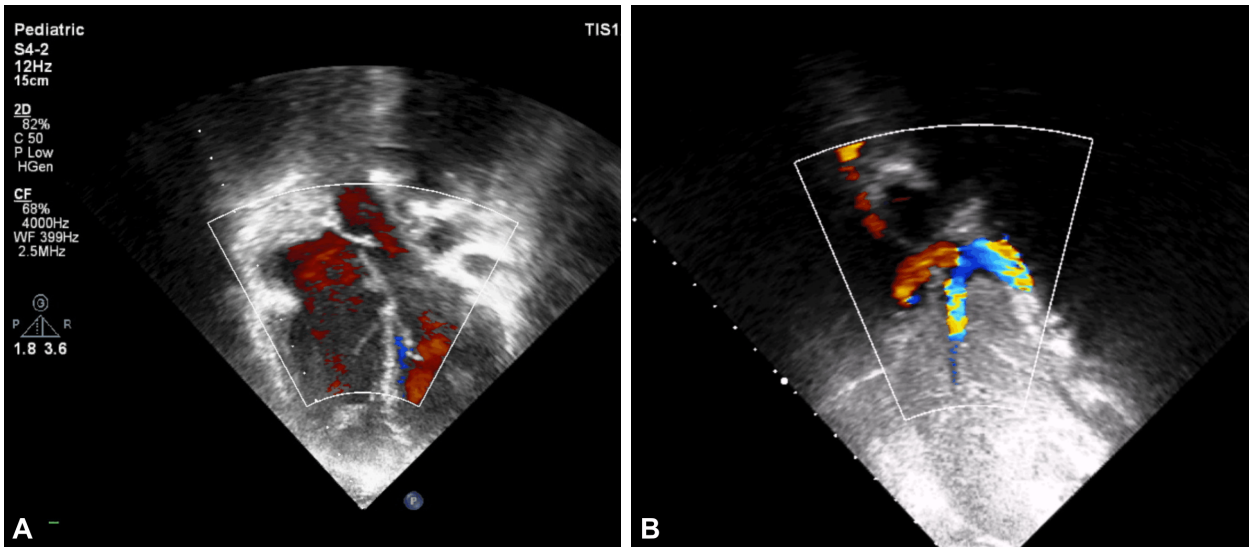

Figure 6 – ***Postoperative echocardiographic images: (A) Four chambers view – normal drainage of the right pulmonary vein into the left atrium, laminar flow; (B) Subcostal bicaval view – the non-obstructive flow of the suprahepatic vein, IVC, and SVC. IVC: Inferior vena cava; SVC: Superior vena cava.***

Discussions

First described in 1836, by Chassinat, SS has an incidence of 1–3/100 000 newborns, representing 0.5–1% of all the congenital heart diseases (CHDs) [1, 5]. It consists of total or partial abnormal pulmonary venous drainage of the right lung into the inferior vena cava, various degrees of a hypoplastic right lung, and of the possible systemic arterial supply to the right lung [6]. The name of the syndrome comes from the resembling of the right pulmonary vein descendent collector to the curved Middle Eastern Ottoman sword called "scimitar".

One of the first largest studies on SS was published in 1956, by Halasz *et al.* They analyzed data from 21 patients underlining the importance of bronchographic and angiographic evaluation [2]. Another large study on SS was reported by Vida *et al.* on 26 patients in a multicentric study. In this study, the median age of the patients at the moment of surgery was 11-year-old (1.8–19.9), but both neonatal and adult cases are described in the literature. Sixty-three percent of the patients in this group [6] had an ASD secundum type and this was the most frequent association. Less frequently, other associations were described: dextrocardia (27%), right lung hypoplasia (27%), ventricular septal defect (3.8%), severe mitral valve regurgitation (3.8%) [6]. Other authors described different association of the SS with CHD, such as tetralogy of Fallot, coarctation of the aorta, hypoplastic aortic arch, patent *ductus arteriosus* (PDA), anomalous origin of the left coronary artery, *truncus arteriosus* [7], anomalies of the systemic venous return (interrupted IVC with azygos vein continuation and the presence of the left superior vena cava) [8, 9], ipsilateral diaphragmatic anomalies, localized bronchiectasis, horseshoe lung, pulmonary

sequestration, genitourinary tract abnormalities [10]. Brown *et al.* published a study on nine patients with SS. None of the patients had ASD, or pulmonary hypertension [11].

Sometimes, ectopic liver into the inferior right lung, associated with SS is misdiagnosed as sequestration and the correct diagnosis is revealed at morphological examination after lobectomy [12].

In our case, the SS was associated with a retroesophageal right subclavian artery, obstructed right bronchus, diaphragmatic hernia with liver tissue, "S"-type thoracic scoliosis, the right ureter duplication, duplication of the left basinet.

Aberrant right subclavian artery (ARSA) is also a very rare congenital vascular malformation – with a prevalence of 0.2% to 13.3% [13] of the general population. ARSA usually has an asymptomatic evolution and does not need a vascular surgery except for cases of "dysphagia lusoria" (swallowing difficulties) or hematemesis.

Clinical manifestations may vary in SS from severe heart failure with pulmonary hypertension to totally asymptomatic. In the study of 26 patients, 73% had symptoms of recurrent pneumonia, upper respiratory tract infections, cardiac failure (15.4%) and 63% had cardiac anomalies associated [6]. Wheezing may be present as a manifestation of possible bronchial anomalies [14]. Hemoptysis is a possible clinical manifestation of the SS, if there is a feeding artery to the pulmonary circulation associated or not with sequestration [9, 15].

Diagnosis of the SS can be suspected on the base of characteristically radiological sign (scimitar vein). Echocardiography, CT and angiography can be helpful in confirming the diagnosis in all its complexity of forms and associations [16]. In the study published by Vida *et al.*, cardiac catheterization was performed in 85% of the patients and magnetic resonance in 11%. Interventional methods are gaining more space in the diagnosis and treatment of CHDs with increasing complexity of the cases [17–19]. Coil embolization may be useful to treat aortopulmonary collaterals, which may be present in 15% of the patients. For all these patients, the treatment is performed preoperatively in all patients. Pulmonary sequestration in the same study was present in 7.7% of the patients [6].

Surgical correction should be considered in the presence of significant left to right shunting and pulmonary hypertension. This involves mainly two possible interventions. The most frequently used is the creation of an interatrial baffle to redirect the pulmonary venous return into the left atrium. In the study performed by Vida *et al.* on 26 patients diagnosed with SS, this intervention was performed in 69% of the patients (group 1). For the other 31% of the patients, the surgical repair consisted into the redirecting the anomalous scimitar vein towards the left atrium (group 2) [6]. The mortality (early and late) was 7.7% (one patient from each group) mainly related to complications of the disease (pulmonary hypertensive crises). As post-operatory complications, Vida *et al.* reported complete occlusion of the scimitar drainage in both groups and this appeared in 16% of the cases. Other patients necessitated either balloon dilatation or stenting of the scimitar vein for scimitar vein stenosis (more in the reimplantation group 25%, comparative to 6% in the baffle creation) [6]. Other complications reported by Vida *et al.* are atrial fibrillation, recurrent respiratory tract infections, pulmonary hypertension and cyanosis [6]. Brown *et al.* performed for all reported patients only direct reimplantation of the scimitar vein to the left atrium without any complications or death [11]. Right pneumonectomy or lobectomy also are mentioned as a last possible therapy [6].

The postoperative outcome depends on systemic arterial supply to the right lung, stenosis of the scimitar vein, and the association with other cardiac anomalies, which may generate irreversible pulmonary hypertension and obstructive pulmonary artery disease [6].

For the presented patient, there is a discrepancy between the scarce clinical presentation and the number of pathologies associated including large ASD, pulmonary hypertension, anomalies of the lungs, aortic arch, diaphragm, liver, spine, kidney. The presence of the ARSA may play a role in the future if an aneurysmal dilation of the ARSA appears in the proximity of the esophagus.

Conclusions

Scimitar syndrome is a rare congenital heart disease, which may vary in clinical presentation, from asymptomatic to severe heart failure associated with pulmonary hypertension patients. Often is associated with other congenital heart diseases or different other anomalies. Complications may appear and they make the management more difficult and increase the mortality. The diagnosis should be straightforward in presence of the characteristic radiological scimitar sign. A complete investigation is necessary in order to establish the best surgical approach according to associations and complications.

Conflict of interests

The authors declare that they have no conflict of interests.

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