Uncommon variants of the scimitar syndrome in two siblings

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ABSTRACT

The Scimitar syndrome is a complex association of cardiovascular and bronchopulmonary abnormalities, with the main feature a partial or total anomalous right pulmonary venous drainage to the inferior vena cava. A number of cases that lack of all the features of the typical syndrome have been described as Scimitar variant, but the incidence is rare. Familial occurrence is exceptional and limited to few cases in literature. We report two sibling diagnosed with an uncommon variant of the Scimitar syndrome.

Keywords: Anomalous pulmonary venous drainage, familial Scimitar syndrome, Scimitar, Scimitar syndrome, Scimitar variant

INTRODUCTION

The Scimitar syndrome is a spectrum of cardiovascular and bronchopulmonary abnormalities. Its classical form is represented by partial or total anomalous pulmonary venous drainage of the right lung to the inferior vena cava, varying degrees of right lung and right pulmonary artery hypoplasia that results in dextroversion of the heart, and an anomalous systemic blood supply from the abdominal aorta to the inferior segments of the right lung. Anomalies of the right bronchial tree are invariable features.

Patients at variance from the classical form but with some features of Scimitar syndrome, cases with the main features located on the left and those in whom some feature of the Scimitar syndrome were present but associated with normal pulmonary venous connections or with partial anomalous pulmonary venous drainage to an uncommon location have been described as "Scimitar variants" although there is still no general agreement about a clear definition of these forms. "Incomplete" or "variant" cases of Scimitar syndrome have been described, as well as the association of the classical form with other cardiac abnormalities.[1-3]

Access this article online

Quick Response Code:

Website: www.annalspc.com

DOI: 10.4103/0974-2069.149523

Familial occurrence of the syndrome is limited to few reports in the literature.[4-7]

We describe two cases of an uncommon variant of Scimitar syndrome diagnosed in two siblings.

CASE REPORT

There was no familial history of congenital heart disease, extra-cardiac malformations, and parents were not consanguineous.

Patient 1

This female had an antenatal diagnosis of Scimitar syndrome at 24 weeks of gestational age. She was born at full term and had an uncomplicated postnatal course. She was asymptomatic at first cardiac assessment at few weeks of age but echocardiogram showed an unexplained mild left ventricular enlargement and dysfunction (fractional shortening 25%). At 6 months the patient underwent cardiac catheterization and bronchoscopy under general anesthesia and this revealed a Scimitar variant consisting of: Severe stenosis of the right pulmonary vein at its connection with the inferior vena cava, atrio-ventricular and ventriculo-arterial concordance, no interatrial communication, with a fistulous communication of the anomalous vein to the right upper pulmonary vein which drained normally to the left atrium [Figure 1]. There was absent right pulmonary artery and right lung hypoplasia, one large aorto-pulmonary collateral arising from the coeliac axis to right middle and lower lobes, dextroposition of the heart and an anomalous left circumflex coronary artery arising from the proximal left pulmonary artery. The pulmonary arterial pressure was normal, 31/11, mean 18 mmHg. Bronchoscopy

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showed an absent right upper bronchus with malacia and hypoplasia of the right middle and lower bronchi. She underwent two transcatheter coil embolisations of the aorto-pulmonary collateral (at age 6 and 24 months).

Because of the left ventricular dysfunction treatment was commenced with an angiotensin-converting-enzyme (ACE) inhibitors and the patient remained symptom free. Left ventricular size and function had returned to normal by the age of 12 months. At 9 years follow-up left ventricular fractional shortening was 36% and end-diastolic dimension 41 mm (Z score + 1.08).

The decision of the team was for conservative approach in view of the good clinical condition of the patient as well as the normalization of the ventricular function.

The initial left ventricular dysfunction can be explained by the anomalous origin of circumflex coronary artery, and the improvement observed with the development of collateral coronary circulation over the years, as well as the effect of ACE inhibitors.

The development of a fistulous communication between the anomalous right lower pulmonary vein and the right upper pulmonary vein draining to left atrium could be interpreted as an error which occurred during embryogenesis, when the splanchnic plexus connected to the lung buds is also related to the left atrium, that regresses lately. Otherwise it could be explained as a mechanism to bypass the subocclusion at the level of the inferior vena cava allowing the blood flow redirection to the left atrium.

Patient 2

This male had an antenatal diagnosis of Scimitar syndrome at 15 weeks of gestational age. He was born at term and remained symptom-free at the first clinical evaluation. Postnatal assessment, including cardiac catheterization, confirmed the diagnosis of Scimitar variant with total anomalous right pulmonary venous drainage to the inferior vena cava below diaphragm, although there was severe obstruction of the anomalous vein at the junction with inferior vena cava; a collateral vessel was seen between the right pulmonary vein and artery through selective injection into the inferior vena cava [Figure 2].

Interestingly, this was initially detected on echocardiogram by retrograde flow seen in the hypoplastic right pulmonary artery. There was also right lung hypoplasia with the medium and lower lobe supplied by two aorto-pulmonary collaterals arising from the descending aorta, above the diaphragm and from coeliac axis [Figure 3]. Transcatheter coil embolization of the collaterals was performed at 3 months and 4 years of age. The patient remains in good condition with only mild wheezing.

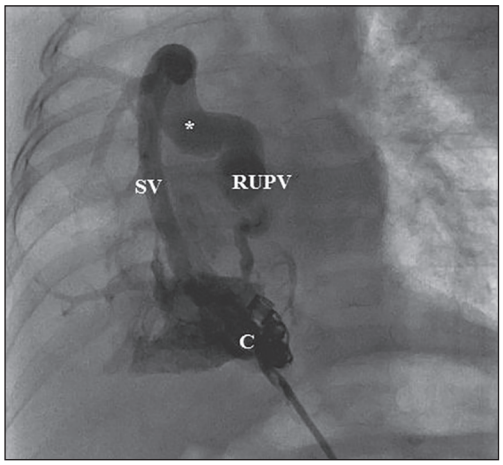

Figure 1: Patient 1, anterior projection of selective angiogram into the aorto-pulmonary collateral during coil embolization (C). There is an anomalous right Scimitar vein (SV) draining to the inferior vena cava (IVC) which was stenosed at the junction with inferior vena cava and connected to the right upper pulmonary vein (RUPV) draining to left atrium by a large and tortuous fistula (*). There is dextroposition of the heart

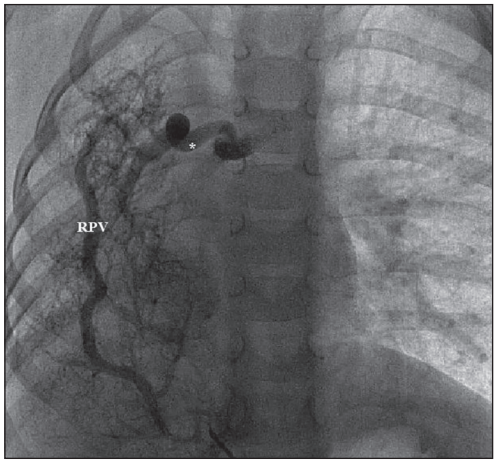

Figure 2: Patient 2, selective injection in IVC showing anomalous drainage of all the right pulmonary veins (RPV) to the hepatic vein with severe pulmonary vein stenosis and a collateral vessel between the right pulmonary vein and artery (*)

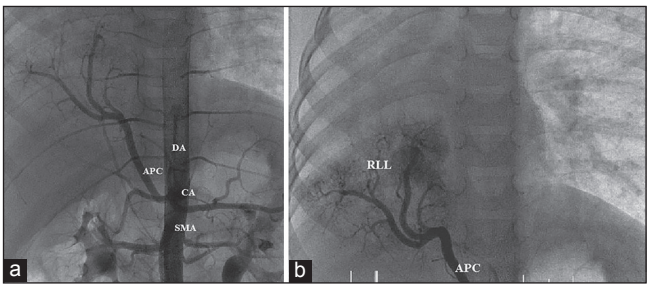

Figure 3: Selective injection into the descending aorta showing a moderate size aorto-pulmonary collateral (APC) arising from Coeliac axis (CA) and supplying the lower part of right lower lobe. SMA = superior mesenteric artery (b) (bis): Selective injection into the descending aorta showing a moderate size aorto-pulmonary collateral (APC) supplying the lower part of the right lower lobe (RLL)

COMMENTS

We report two cases of rare variants of the Scimitar syndrome in the same family.

The presence of a Scimitar variant with absence of right pulmonary artery, associated with an anomalous origin of circumflex coronary artery from pulmonary trunk has not been reported so far. The presence of anastomotic connections between the Scimitar vein and left atrium has been described previously in classical form of Scimitar syndrome, although reported in about 10% of the population of a large series of patients;[8] we are unaware of previous descriptions of a fistulous communication between the Scimitar vein and right pulmonary artery, seen in patient 2.

Despite severe stenosis of the right anomalous pulmonary vein at the junction with inferior vena cava, both patients remained well during follow up; this can be explained by the presence of the anastomosis of that venous system with the normally draining right upper pulmonary vein in case 1, that permits the physiologic drainage of arterial blood from the right lung to the left atrium.

Furthermore the presence of severe lung hypoplasia together with right pulmonary artery hypoplasia or absence may account for the lack of symptoms of pulmonary hypertension.

Previous familial occurrence of Scimitar syndrome and variants have been described as well as recurrence of pulmonary sequestration[3,9] and other bronchopulmonary abnormalities forming part of the so-called "bronchopulmonary foregut malformation complex".[10]

Ruggieri *et al.*[6] described two siblings presenting with infantile form of Scimitar syndrome with partial right anomalous pulmonary venous drainage to the inferior vena cava, hypoplastic right pulmonary artery and lung, aorto-pulmonary collateral to the right lung and secundum atrial septal defect associated to craniofacial and central nervous system (CNS) anomalies.

A genetic explanation for this occurrence has not been determined yet, but familial cases of Scimitar syndrome, despite extremely uncommon, could be the model for future investigations.

In conclusion the Scimitar syndrome includes a spectrum of variants whose morphological characterization can be challenging. The identification of all the features may require a combination of complementary investigations. The pathophysiology of such lesions is still unclear, but one hypothesis speculates an embryogenic error in early development or a compensatory mechanism to the distal obstruction at the level of inferior vena cava.

The therapeutic approach should be tailored for each individual case, and should also consider the clinical status, the complex anatomy and the treatment options.

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How to cite this article: Bo I, Daubeney PE, Rigby ML. Uncommon variants of the scimitar syndrome in two siblings. Ann Pediatr Card 2015;8:56-8.

Source of Support: Nil, **Conflict of Interest:** None declared