A Scimitar Syndrome Variant Associated with Critical Aortic Coarctation in a Newborn

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Patient: Female, newborn
Final Diagnosis: Scimitar syndrome variant associated with critical aortic coarctation
Symptoms: Absence of pedal pulse • apnea • cardiogenic shock • extremely poor clinical condition • hepatomegaly • livedo reticularis • pallor • tachycardia
Medication: —
Clinical Procedure: —
Specialty: Cardiology • Pulmonology

Objective: Rare disease
Background: Scimitar syndrome (SCS) is a rare congenital cardiopulmonary malformation, characterized by anomalous pulmonary venous drainage from the right lung associated with aortopulmonary collateral arteries and pulmonary hypoplasia. The variant described in the case presented here, with total anomalous right pulmonary venous drainage into the superior and inferior vena cava, can be expected in 2% of patients with scimitar syndrome. To the best of our knowledge, the association between the variant of SCS and coarctation of aorta described in our patient has never been reported before in the literature.

Case Report: A female newborn with a gestational age of 35 weeks presented with a rare combination of scimitar syndrome and aortic coarctation. The patient had a variant of SCS that included anomalous drainage of the right upper and lower pulmonary vein into the superior and inferior vena cava, respectively; relative right lung hypoplasia; and right lower lobe sequestration supplied by aortopulmonary collateral arteries that originated from the truncus coeliacus. The diagnosis was confirmed with computed tomography angiography after resection of the aortic coarctation with extended end-to-end anastomosis. Subsequently, interventional closure of the collateral artery supplying the right lower lobe was performed with an AMPLATZER™ Vascular Plug 4. The patient’s clinical course was complicated by suspicious acute endocarditis and chylous pleural effusion. After a prolonged hospitalization, she was discharged in clinically stable condition.

Conclusions: The diagnosis of SCS should be considered when pulmonary hypertension persists after coarctation repair in a child with dextroposition of the heart and right lung hypoplasia. Successful treatment of this rare combination of conditions calls for teamwork by highly experienced specialists.

MeSH Keywords: Aortic Coarctation • Persistent Fetal Circulation Syndrome • Premature Birth • Scimitar Syndrome

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Background

Scimitar syndrome (SCS) is a rare congenital malformation, characterized by partial or total anomalous pulmonary venous return from the right lung, pulmonary hypoplasia, and aortopulmonary collateral arteries [1–5]. Pulmonary hypertension is a common finding and a poor prognostic sign. The combination of SCS and aortic coarctation is extremely rare, and only a few cases have been described in the literature [6]. With this combination, clinical manifestations at birth vary and can range from no symptoms to respiratory distress and heart failure. The prognosis depends on the extent of pulmonary hypoplasia, the degree of pulmonary hypertension, and the outcomes of cardiac surgery and interventional procedures [1–5].

In this report, we discuss the case of a female newborn with an unusual association between an SCS variant and aortic coarctation, which resulted in challenging diagnostic observations and a complex clinical course.

Case Report

The patient was the third child of a 33-year-old, non-smoking, healthy mother who was regularly monitored during her pregnancy. Amniocentesis showed no genetic abnormalities, including DiGeorge syndrome. A morphological ultrasound scan performed at 24 weeks' gestation demonstrated intrauterine growth restriction (IUGR) and oligohydramnios. The mother was hospitalized at 28 weeks' gestation for corticosteroid treatment. Fetal echocardiography showed an enlarged right ventricle and pericardial effusion. Because of the IUGR, delivery was induced at 36 weeks' gestation. The newborn weighed 2040 g and had Apgar scores of 8 at 1 and 5 minutes. The early postnatal period was uneventful and the newborn was discharged from the maternity hospital in good clinical condition 14 days after birth.

At age 21 days, the newborn was examined and found to be in extremely poor clinical condition, with signs of cardiogenic shock, tachycardia, hepatomegaly, pallor, mottled skin, absence of peripheral pulse, prolonged capillary refill (over 6 seconds) and periodic apnea. A chest X-ray showed cardiomegaly, dextroposition of the heart; and relative right lung hypoplasia (Figure 1). Echocardiography revealed a 4-mm hypoplastic left aortic arch, aortic coarctation with narrowing of the isthmus to 1.5 mm and a posterior shelf (Figure 2), closed ductus arteriosus, bicuspid aortic valve, enlarged right ventricle, decreased systolic function in both ventricles, mild pulmonary hypertension, and mild pericardial effusion. The left pulmonary veins drained into the small left atrium near the septal region, while there were anomalous retrograde venous drainage into the inferior vena cava and no connection between the right pulmonary veins and the left atrium (Figure 3). The newborn was intubated and treated with prostaglandin E and cardiotonic and decongestive agents. Because of her poor clinical condition, oliguria, and increased lactate levels, resection of the coarctation segment and extended end-to-end anastomosis was performed a few hours later.

Soon after the surgery, the newborn's clinical condition and echocardiography findings improved (Figure 4). Because of persistent pulmonary hypertension, sildenafil was administered. To evaluate pulmonary venous drainage, a computed tomography angiography was performed. Congenital pulmonary venolobar syndrome (scimitar syndrome) was diagnosed, which consisted of anomalous drainage of the right upper pulmonary vein into the superior vena cava and of the right lower pulmonary vein into the inferior vena cava; right lung hypoplasia; dextroposition of the heart; and intrapulmonary sequestration in the right lower lobe supplied by the systemic artery from the celiac trunk (Figures 5, 6). When the patient was age 5 weeks, interventional catheterization was performed in coordination with the referring center (Motol University Hospital, Prague, Czech Republic) and showed a mean pressure in the pulmonary artery of 27 mmHg. The aortopulmonary collateral artery then was occluded completely with an AMPLATZER™ Vascular Plug 4. After the procedure, chylous pleural effusion was noted around the left lung and the pleura was drained. Prolonged treatment with vancomycin and cefotaxime was administered after the patient's blood culture showed Staphylococcus epidermidis and infective endocarditis was suspected because she had an indwelling catheter and increased levels of C-reactive protein. The patient was discharged at age 4 months in clinically good condition with no pulmonary hypertension. Her clinical course was uneventful at 1-year follow-up.

Figure 1. Chest X-ray showing cardiomegaly, displacement of the heart to the right, and relative right lung hypoplasia with abnormal opacity.
Figure 2. Echocardiogram of the aortic arch before surgery. (A) Narrowing of the aortic isthmus. (B) Turbulent flow. (C) Coarctation flow pattern on pulsed-wave Doppler. (D) Continuous-wave Doppler with diastolic runoff.

Figure 3. Echocardiographic 4-chamber view. (A) Displacement of the communication between the left pulmonary veins (LPV) and left atrium. (B) The relationship between the left atrium (LA) and the left ventricle (LV). There is no communication between the right pulmonary veins and the left atrium. RA – right atrium, RV – right ventricle.
Figure 4. (A–D) Echocardiogram of the aortic arch after surgery showing improvement and normalization of the findings illustrated in Figure 2.

Figure 5. Computed tomography angiography. (A) Dextroposition of the heart, relative right lung hypoplasia, and displaced communication between the left pulmonary veins (LPV) and left atrium (LA). (B) Relatively hypoplastic right pulmonary artery (RPA). LV – left ventricle, RA – right atrium, RV – right ventricle.
Figure 6. Computed tomography angiography demonstrating a variant of scimitar syndrome. The white arrows point to right upper and right lower pulmonary vein drainage into the superior and inferior vena cava, respectively. The black arrow points to an aortopulmonary collateral artery running from the celiac trunk toward the right lower lobe of the lung.

Discussion

The incidence of SCS is 1 to 3 per 100,000 live births and its clinical presentation in children is variable [1]. Up to half of patients with it are symptomatic and present within the first 6 months of life [7]. The anomalies most commonly associated with SCS are atrial septal defect, ventricular septal defect, patent ductus arteriosus, anomalous origin of the coronary artery from the pulmonary trunk, persistent left superior vena cava drainage into the coronary sinus, and coarctation of the aorta [7–9]. Symptomatic infants with SCS typically have associated anomalies, and aortic coarctation can be expected in 10% of them. Treatment of SCS includes conservative therapy, surgical repair, occlusion of collateral arteries, pneumonectomy, and lung transplantation. Up to one-third of infants with SCS who are symptomatic die. In the case presented here, we decided to perform interventional embolization of the aortopulmonary collateral artery because the patient was on prolonged mechanical ventilation; the result was optimal.

Half of patients who are diagnosed with SCS after infancy remain asymptomatic, and the condition is an incidental finding, based on seeing the characteristic “Turkish sword” in the right lung on chest X-ray. Approximately 18% of patients with SCS have variants that differ from the classical appearance (anomalous drainage of the right pulmonary vein into the inferior vena cava and aortopulmonary collateral arteries in the right lung) [8]. The variant described in our patient, with anomalous total right pulmonary venous drainage into the superior and inferior vena cava, can be expected in 2% of cases of SCS.

Diagnosis of critical congenital heart disease (CHD) is based on 3 screening methods: prenatal ultrasound, clinical examination after birth, and pulse oximetry. In Slovenia, 90% of heart anomalies are diagnosed in timely fashion using these 3 methods [10]. In our case, all 3 screening methods were applied, but the patient’s CHD was not diagnosed. Left-sided, obstructive CHD is known to present the greatest screening challenge, and sometimes it cannot be diagnosed in the first days of life. Normal hemodynamic processes result in closure of the ductus arteriosus between the second and fourth weeks after birth, which can lead to severe heart failure or even death [10]. Timely diagnosis of critical heart anomalies in newborns is of great importance because it reduces mortality, and additional screening and pulse oximetry at age 2 to 3 weeks may be warranted. In our case, closure of the ductus arteriosus on the basis of aortic coarctation and scimitar syndrome led to severe heart failure and a life-threatening situation. The patient’s clinical condition was worsened by pulmonary hypertension, which could have been related to the coarctation and SCS-related pulmonary hypoplasia. However, because our patient was in such poor condition, it was not possible to evaluate the SCS variant before the aortic surgery. We concluded that hemodynamic compromise and pulmonary hypertension were mainly associated with the critical coarctation and decided to postpone a complete diagnostic evaluation of the right pulmonary venous drainage until after surgery.

Conclusions

SCS with critical aortic coarctation is a very rare form of CHD in neonates. An association of the variant of SCS and coarctation of the aorta as described in our patient has never been reported in the literature before. In general, presentation of SCS in infancy with heart failure, pulmonary hypoplasia, and hypertension is associated with a poor prognosis [6]. A diagnosis of SCS should be considered when pulmonary hypertension persists after coarctation repair in a child with dextroposition of the heart and right lung hypoplasia. Successful treatment of this rare combination calls for teamwork by highly experienced specialists.
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