Infantile scimitar syndrome with unusual associations

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ABSTRACT

Scimitar syndrome is a variant of partial anomalous pulmonary venous connection (PAPVC), in which all or part of the right lung is drained by right pulmonary veins that anomalously connect to the inferior vena cava (IVC). The affected lung and its associated airways are often hypoplastic. In addition, aortopulmonary collateral vessels may be involved on the affected side, causing sequestration of that side; such involvement is commonly associated with cardiac defects. We report a case of infantile scimitar syndrome that involved a typical association with the right lung, but with extremely unusual associations with congenital hydrocephalus and heart blockage. The presentation of this case and the role of different diagnostic approaches and management are discussed.

Saudi Med J 2017; Vol. 38 (7): 764-767
doi: 10.15537/smj.2017.7.18365

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Received 10th January 2017. Accepted 7th June 2017.

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Disclosure. Authors have no conflict of interests, and the work was not supported or funded by any drug company. Dr. Adel S. AlHarbi is a member of the Editorial Team, and was therefore excluded from any final editorial decisions regarding this paper.

Case Report. Patient information. A newborn Saudi male antenatally diagnosed with congenital hydrocephalus developed mild respiratory distress soon after delivery.

Clinical finding. On examination, he had marked decreased breath sound on right sided chest. Head circumference was within normal range, loud heart sound in the pulmonary area. To treat this distress, nasal continuous positive airway pressure (NCPAP) was administered for respiratory support. The medical history and final resolution of the case (over a month period) was summarize in Figure 1.

Diagnostic assessment. A chest x-ray showed right lung collapse with a rim of pneumothorax on the left side. A chest CT scan performed after intravenous contrast (Figure 2) with CT angiography showed a hypoplastic right pulmonary artery and large feeding vessels supplying the lower lobe; these findings were consistent with sequestration of the right lower lobe. Hypoplasia of the right lung and airspace disease involving the right lower lobe are features of scimitar syndrome. An ECG revealed complete heart blockage,
and his heart rate remained between 70 and 100 beats per minute; negative findings were obtained for maternal antibodies (anti-Ro [SSA] and anti-La [SSB] antibodies). Echocardiography showed a large ventricular septal defect (VSD) and a small patent ductus arteriosus (PDA). Both of these features shunted bidirectionally, indicating equal pulmonary and systemic arterial pressures.

**Therapeutic intervention.** His condition was managed conservatively. He underwent cardiac catheterization to close the PDA and collateral vessels. Angiography confirmed the diagnosis and showed a tiny PDA with almost no shunt. A collateral vessel that fed the lower right lobe was extremely narrow and inaccessible. Repeated angiography showed stenosed vessels due to repeated access attempts.

A follow-up cardiology appointment was scheduled after discharge, and cardiac catheterization was repeated one year later in the location where the feeding vessel from the abdominal aorta was coiled (Figure 3). The pulmonary venous return of the entire right lung drained through the anomalous vein to the inferior vena cava (IVC). Fortunately, the patient continued to suffer from cardiorespiratory failure and eventually passed away.

Figure 1 - Timeline summary of the case.

![Timeline summary of the case](image-url)
Infantile scimitar syndrome with unusual associations ... Al-Shamrani et al

Figure 2 - A coronal view produced by chest CT, showing a sequestrated right lower lobe with a large feeding vessel (arrow) from the abdominal aorta and a hypoplastic right lung.

Figure 3 - AP projection showing the positioned coil occluding the arterial feeding vessel (arrow) and a venous catheter passing from the IVC to the lower right anomalous scimitar pulmonary vein.

Discussion. Scimitar syndrome exhibits autosomal dominant inheritance with variable penetrance.3 The median age for presentation of this syndrome is 7 months, although the age at presentation varies.3 Many patients are asymptomatic throughout childhood and typically present with recurrent respiratory tract infections.3 The severity of scimitar syndrome varies considerably. Some patients with the relevant anomaly are completely unaware of their condition or present with mild respiratory symptoms that neither affect their lives nor require intervention. However, this defect can potentially cause significant health problems or endanger the heart;4 such issues were observed in our case, in which infantile scimitar syndrome was diagnosed. In this case, pulmonary pressure was equivalent to systemic pressure, with bidirectional shunts through the PDA. We have described the second reported case involving the association of scimitar syndrome with congenital hydrocephalus,5 which further adversely affected the outcome. This association appears to be coincidental because both anomalies could concurrently arise in early embryogenesis and because the neonate exhibited no apparent cause of hydrocephalus such as infection or hemorrhage. The “scimitar sign” on chest x-rays was first described by Godwin et al,6 although this sign is an important diagnostic finding, it is not necessarily specific. Doppler examinations may show the union of the scimitar and systemic veins. The CT and MRI permit direct visualization of the anomalous vein, and angiographic techniques and multiplanar reconstructions allow radiologists to determine arterial and bronchial anatomy in detail.7 Conventional angiographic studies may still be ordered by surgeons to delineate the arterial and venous anatomy prior to surgical repair, and virtual bronchoscopy may show the fish-mouth appearance of the affected side and variable degrees of airway narrowing.7 Echocardiography could potentially be used to determine whether common cardiac defects such as atrial septal defect (ASD) and PDA are present and whether significant pulmonary hypertension and drainage of the pulmonary veins exist. Bronchoscopy can be utilized in cases of scimitar syndrome to assess the presence of airway hypoplasia.8 Because of the wide clinical spectrum for scimitar syndrome, the strategy for medical intervention depends on the severity of presentation and the amount of blood flowing to the IVC from completely or partially anomalous pulmonary veins. If there is a small amount of drainage, therapy may not be required. Management is often supportive and can include the prescription of cardiac medication if volume overload exists; the prescription of antibiotics for chest infections; the promotion of good nutrition; oxygen supplementation and the prescription of sildenafil for pulmonary
hypertension; and ventilatory support which all were utilized in our patient.4 The presence of respiratory symptoms is typically one of the main indications for surgical correction.3,9 Furthermore, surgical repair seldom results in normal blood flow to the right lung and abolishes the increased left-to-right shunt with pulmonary volume overload that often results in improving clinical conditions in most patients.9,10 Simple ligation or coil embolization of abnormal arterial vessels has been advocated as the best and simplest form of treatment, particularly in symptomatic infants as in our case.9,10 In contrast, Huddleston et al9 recommended repair of the anomalous venous return and the ligation of collaterals in symptomatic patients.9 Thrombosis and fibrosis of the redirected pulmonary veins are serious complications of the surgical re-implantation procedure, which often requires rethoracotomy with resection of the remaining lung. Pneumonectomy (either as a primary therapy or after repair failure) has produced similar results.3,10

In conclusion, scimitar syndrome is a rare disease that has been well described in the literature, but has variable presentations and associations; thus, a great deal of caution is required with respect to diagnose infantile scimitar syndrome. This condition can be initially suspected from a chest x-ray, but is typically confirmed via CT angiography. For scimitar syndrome that presents in infants, large blood flow into the IVC from the anomalous pulmonary veins and the presence of hydrocephalus are negative prognostic factors.

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