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SCIMITAR SYNDROME: UNVEILING THE COMPLEXITIES OF A RARE CONGENITAL CARDIOPULMONARY ANOMALY

Nargis Mateen, Abraam Rezkalla, Nagihan Orhun

Department of Internal Medicine, St. Josephs University Medical Center, Paterson, USA

Corresponding author's e-mail: r_mateenn@sjhmc.org

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ABSTRACT

Scimitar syndrome is a rare congenital anomaly characterised by anomalous pulmonary venous drainage of the right lung into the inferior vena cava, often accompanied by right lung hypoplasia and dextroposition of the heart. Here, we present the case of a 24-year-old female with scimitar syndrome and a history of asthma who presented with acute exacerbation of asthma. A chest X-ray revealed opacification of the right hemithorax, and a CT angiography revealed congenital hypoplasia of the right lung and dextrocardia. This report provides a detailed description of scimitar syndrome to spread awareness of this rare diagnosis and highlights the unique and various constellations of anatomical abnormalities seen in scimitar syndrome that make diagnosis difficult.

KEYWORDS

Scimitar syndrome, pulmonary hypoplasia, asthma exacerbation, dextrocardia, rare congenital anomaly

LEARNING POINTS

- Scimitar syndrome is an extremely rare congenital cardiopulmonary anomaly and diagnostic challenge, with key features of partial or complete anomalous venous drainage of the right lung into the inferior vena cava. It exhibits varying degrees of right lung and pulmonary artery hypoplasia, dextrocardia and abnormal arterial supply to the ipsilateral lung.
- The adult form of this rare syndrome may remain underdiagnosed for years due to its less severe clinical presentation or incidental findings, such as recurrent pulmonary infections or unexplained dyspnoea.

INTRODUCTION

Scimitar syndrome was first described by George Cooper in 1836, characterised by a complex constellation of cardiopulmonary anatomical abnormalities. The prevalence of scimitar syndrome is estimated to be 1-3 per 100,000 births, highlighting its rarity and diagnostic challenges due to its heterogeneous clinical presentation[1]. Scimitar syndrome has a variable presentation, ranging from patients that are asymptomatic to patients with signs of congestive heart failure and/or respiratory distress[2]. Due to the rare nature of this syndrome and its wide spectrum of presentations, literature is limited. This case describes a 24-year-old female with scimitar syndrome who underwent a pneumonectomy at 6 months of age, who presented with recurrent acute asthma

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exacerbations. It aims to shed light on the complexities and challenges of a rare cardiopulmonary anomaly.

CASE DESCRIPTION

A 24-year-old female presented to the emergency department (ED) with the complaint of worsening shortness of breath. She reported that her symptoms began suddenly on the evening of her presentation and were associated with a dry cough. She denied any instigating factor, fever, chills or exposure to sick contacts. Upon further questioning, the patient reported a history of asthma with multiple exacerbations requiring admission in the past and intubation on one occasion. She denied using any daily inhalers. She also revealed a history of dextrocardia and right lung hypoplasia status post pneumonectomy at 6 months of age.

In the ED, the patient's initial triage vitals were as follows: respiratory rate of 20, oxygen saturation of 96% on 5 litres of oxygen via nasal cannula, blood pressure of 118/87, heart rate of 96 and temperature of 36.2°C. On assessment, she reported having chest tightness, and severely decreased right-sided air entry was found on physical examination. A chest X-ray was then obtained, revealing significant opacification of the right hemithorax with volume loss and a clear left lung, consistent with the patient's history of pulmonary hypoplasia and pneumonectomy. A CT angiography was also obtained to rule out a pulmonary embolism, which revealed congenital hypoplasia of the right lung with marked ipsilateral mediastinal shift and non-visualisation of the right pulmonary artery and its branches. Compensatory hypertrophy of the left lung was also seen, and the heart appeared to be rotated towards the right *(Fig. 1 and Fig. 2)*.

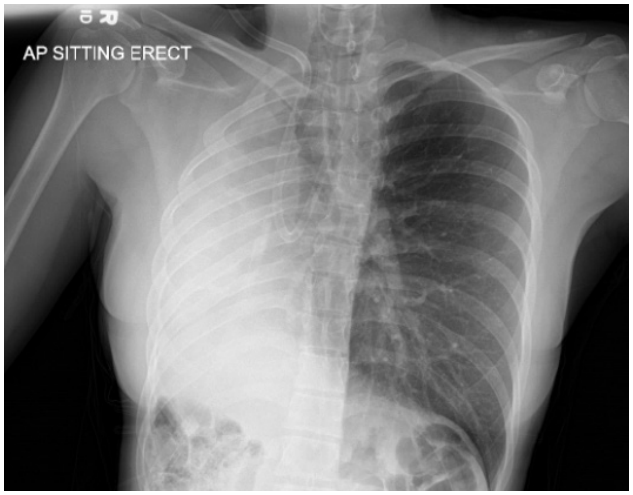


Figure 1. Chest X-ray showing complete opacification of the right hemithorax and a clear right lung.

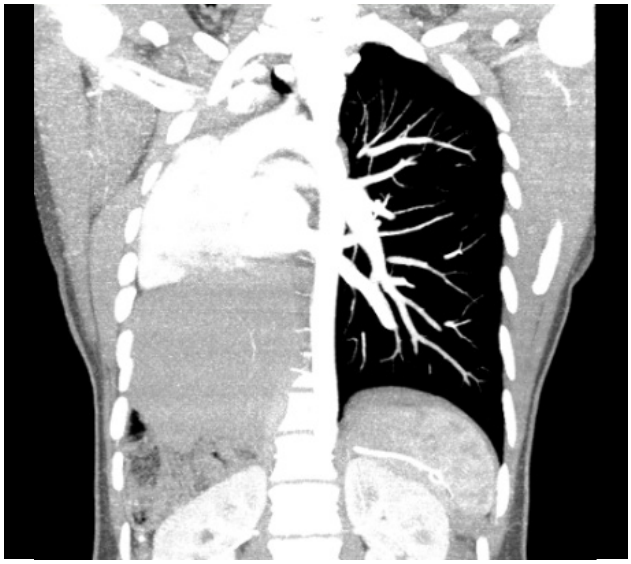


Figure 2. CT angiography of the chest showing congenital hypoplasia of the right lung and dextrocardia. The right pulmonary artery and its branches cannot be identified.

The patient was placed on bilevel positive airway pressure, administered albuterol 10 mg nebulised solution and dexamethasone 10 mg intravenously; however, she continued to report shortness of breath and was subsequently switched to high-flow nasal cannula at 50 litres/min with 40% fraction of inspired oxygen. She was admitted to the medical intensive care unit for acute hypoxic respiratory failure in the setting of asthma exacerbation. Methylprednisolone 40 mg intravenously every 8 hours and albuterol nebulisation were initiated and a respiratory panel, pneumonia panel and COVID-19 test were obtained, which were negative. A 2D echo was also obtained and revealed a normal left ventricular ejection fraction of 55–60%, with no pulmonary hypertension. The patient was later transitioned to nasal canula 3l and shortly after to room air saturating 96%. Pulmonology was consulted and deemed the patient stable for discharge on a prednisone taper, and fluticasone furoate/vilanterol 100 µg - 25 µg/inh and albuterol 90 µg/inh inhalers, with instructions to follow up in the outpatient setting for pulmonary function tests. The patient has since had four more episodes of ED visits over one year for acute asthma exacerbations.

DISCUSSION

Scimitar syndrome is a rare congenital cardiopulmonary defect described by partial abnormal venous connections of the right lower pulmonary vein draining into the inferior vena cava, instead of the left atrium. The characteristic features of this syndrome are a combination of a partial or complete anomalous curved venous drainage of the right lung into the inferior vena cava; variable right lung and pulmonary artery hypoplasia; dextroposition of the heart; and an anomalous systemic blood supply to the ipsilateral lung[2]. The severity of this syndrome is related to the severity of the anatomical abnormalities, the age at which the patient was diagnosed and other existing congenital disorders. Because of the wide range of symptoms at clinical onset, the diagnosis may be difficult, especially in children and young adults with concomitant congenital heart lesions[3].

Dupuis et al. divided scimitar syndrome into three main forms: an infantile form with symptoms and pulmonary hypertension, an 'older' adult form distinguished by being asymptomatic in infancy and a form with associated

congenital cardiac anomalies (most commonly an atrial septal defect)[4]. The infantile form has shown to be associated with comorbidities that include congenital defects such as horseshoe kidney, accessory diaphragm, pericardial absence, phrenic cyst, and eventration or partial absence of the diaphragm. The infantile form can be complicated by a left-to-right shunt, and severe pulmonary hypertension leading to congestive heart failure and a greater mortality risk[2]. Patients may also present with recurrent pulmonary infections especially in the right lower lobe or bronchiectasis, and interstitial pulmonary disorders. The childhood/adult form is usually milder and patients frequently present with an incidental finding of unexplained right heart dilation or recurrent pulmonary infections, or during evaluation of unexplained dyspnoea. However, due to the rarity of the syndrome, many patients remain misdiagnosed for many years[2].

The most common radiographic findings in scimitar syndrome include heart dextroposition and right-sided mediastinal shift with compensatory left lung hyperinflation[5]. The scimitar sign is described as the anomalous vein draining into the inferior vena cava. This sign is the main feature of the syndrome, but it is commonly hidden due to the degree of dextroposition. For this reason, other imaging techniques such as echocardiogram and CT angiogram are required. The CT scans help expose lung sequestration, which is an important finding because it increases the risk for recurrent lung infections. Cardiac catheterisation is occasionally required to confirm the presence of pulmonary hypertension[5].

Scimitar syndrome always requires a multidisciplinary approach. With the infantile form that usually presents with signs of heart failure, medical therapy should be initiated immediately, allowing the patient to grow until further interventions become necessary[2]. A catheter-based approach during angiography consists of eliminating the abnormal systemic arteries from the aorta to the right lung, which allows for clinical improvement by reducing the degree of shunt and decreasing the pulmonary arterial pressures. However, this may not work if the patient also has cardiac defects, which require early surgical intervention. The childhood/adult form requires surgical intervention with symptomatic patients or when the pulmonary to systemic flow ratio is greater than 1.5 in asymptomatic patients[2]. Surgical management consists of redirecting the pulmonary venous drainage into the left atrium. This can be done either via a tunnel in what is known as a 'baffle repair', or transecting the scimitar vein near its entrance into the inferior vena cava and then reimplanting it directly into the left atrium[3]. Repair of the anomalous venous return is generally recommended; however, in symptomatic patients with repeated pulmonary infections, haemorrhage or marked hypoplasia of the right lung, a right lung lobectomy or pneumonectomy is an alternative option leading to similar results[2,3].

In this case, the patient presented with a history of recurrent asthma exacerbations, congenital right lung hypoplasia and dextrocardia. While it was unclear whether the patient had a history of recurrent pulmonary infections, she did report a history of pneumonectomy at 6 months of age. The patient reported recurrent asthma exacerbations and continued to visit the ED for asthma exacerbations following treatment of her asthma. This may be due in part to the significantly reduced total lung capacity in this patient after the pneumonectomy, which can make managing asthma symptoms challenging.

Historically, the prognosis of scimitar syndrome was grim in the infant form. Nowadays, with early diagnosis and an established surgical strategy, the outcomes tend to be good with a low rate of morbidity and mortality after corrective surgery[2].

CONCLUSION

This report presents the case of a patient with scimitar syndrome and recurrent acute asthma exacerbations. This illuminates the diagnostic challenges posed by the syndrome's diverse clinical presentations and provides a comprehensive overview of scimitar syndrome to increase awareness of a rare diagnosis. Despite these challenges, continued research and clinical vigilance are key in advancing our understanding and therapeutic strategies for this complex congenital condition.

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