Cardiology in the Young

cambridge.org/cty

Diaphragmatic eventration as a first sign of Scimitar syndrome

Miguel Vieira Martins[1], Duarte S Martins[2] and Graça Oliveira[3]

[1]Pediatrics Department, Centro Hospitalar Universitario Cova da Beira, Covilha, Portugal; [2]Pediatric Cardiology Department, Hospital Santa Cruz, Centro Hospitalar Lisboa Ocidental, Carnaxide, Portugal and [3]Neonatology Department, Hospital Santa Maria, Centro Hospitalar Universitário Lisboa Norte, Lisboa, Portugal

Abstract

Diaphragmatic eventration is an anomaly of the diaphragm. In Scimitar syndrome, a curved-shaped anomalous pulmonary venous drainage is seen. Association between these conditions is rare. We present a newborn with diaphragmatic eventration, whose diagnosis of Scimitar syndrome was made after surgical repair. Scimitar syndrome is a congenital disorder often associated with other heart and lungs anomalies. Diagnosis can be fortuitous but with important prognostic features.

Brief Report

Cite this article: Vieira Martins M, Martins DS, and Oliveira G (2021) Diaphragmatic eventration as a first sign of Scimitar syndrome. *Cardiology in the Young* **31**: 1870–1872. doi: 10.1017/S1047951121001694

Received: 29 November 2020
Revised: 14 February 2021
Accepted: 9 April 2021
First published online: 4 May 2021

Keywords:
Congenital diaphragmatic defects; Scimitar syndrome; Newborn

Author for correspondence: Miguel Vieira Martins, M.D., Pediatrics Department, Centro Hospitalar Universitario Cova da Beira, Quinta do Alvito, 6200-251 Covilha, Castelo Branco, Portugal. Tel: +351 275 330 000. E-mail:miguelmartins@campus.ul.pt

Background

Diaphragmatic eventration is a congenital anomaly of the diaphragm that arises from poor development or paralysis of the diaphragmatic musculature. This abnormality can easily be mistaken by a congenital diaphragmatic hernia due to its difficult recognition in utero.[1] Surgical repair by diaphragm plication usually benefits significantly symptomatic patients.[2] Moreover, a rare association of diaphragmatic eventration and anomalous pulmonary venous drainage return to the inferior vena cava has been described as a variant of Scimitar syndrome.[3] Scimitar syndrome is a complex congenital anomaly that commonly features a hypogenetic right lung with dextroposition of the heart, anomalous systemic arterial supply to the ipsilateral lung, and a typical curved anomalous right pulmonary vein that drains into the inferior vena cava. The latter resembles a curved Turkish sword named "scimitar", hence its terminology.[4]

Furthermore, it is frequently associated with another rare bronchopulmonary anomaly called horseshoe lung, in which the posterobasal portions of both lungs are fused by a narrow isthmus of pulmonary parenchyma.[5] The annual incidence of Scimitar syndrome can vary from 1 to 3 in 100,000 live births with a 2:1 female predominance. We present a rare case of a term newborn with DE, whose diagnosis of Scimitar syndrome and horseshoe lung was made after surgical repair.

Case report

A full-term female newborn with regular prenatal follow-up had a vaginal delivery with an appropriate weight for gestational age and no resuscitation. At 14 hours, postpartum acute respiratory distress and hypoxemia were developed. Mechanical ventilation was initiated. Chest radiography showed an extensive homogenous hypotransparency of the lower half of the right hemithorax. CT scan was unclear on whether an eventration of the diaphragm or a CDH was present (Fig 1a, black arrow). A fortuitous finding of an abnormal pulmonary venous drainage was also seen in the right lung (Fig 1a, white arrow). After surgical review, further investigation by pulmonary ultrasound at day 4 of life was highly suggestive of DE given the integrity of the diaphragm. At day 5 of life, a surgical diaphragmatic plication was performed with no complications. Mechanical ventilation remained until day 10, and supplementary oxygen weaned until day 18. A follow-up transthoracic heart ultrasound confirmed the anomalous pulmonary vein drainage to the inferior vena cava (Fig 2, arrow) with no evidence of pulmonary hypertension. This finding was corroborated by CT angiography (Fig 1b, black arrow) showing hypoplastic right pulmonary artery and abnormal venous drainage into the inferior vena cava. Furthermore, a horseshoe/pseudo-horseshoe lung was also noticed with a strip of right lung parenchyma that reached the contralateral left lung, posterior to the heart and anterior to the oesophagus.

Blood work showed normal renal and thyroid functions. On abdominal ultrasound, no renal abnormalities were found, but an uterine duplicity was reported. Genetic profiling by *array comparative genomic hybridisation* test was performed with no abnormal results. At day 28 of life, the patient was discharged from the Neonatal ICU. Follow-up consultations were planned to keep a close surveillance with cardiology intervention to be considered should the patient present new symptoms.

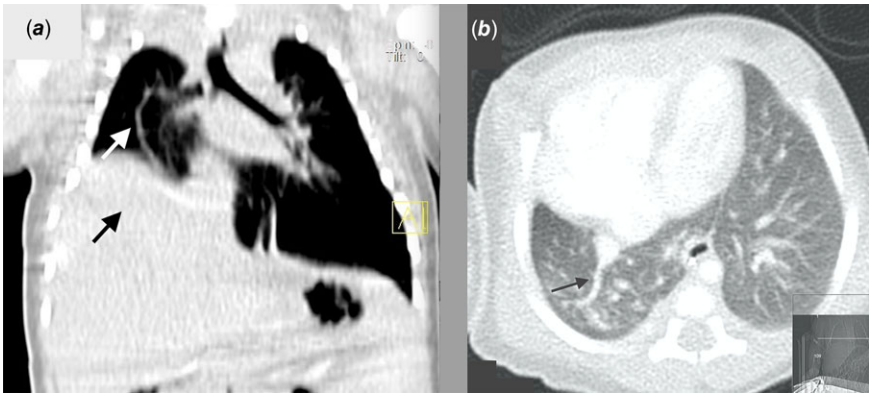

Figure 1. (a) Pre-surgical CT scan in coronal view showing an extensive homogenous hypodensity of the lower half of the right hemithorax (black arrow) and curved sword vascular sign – scimitar vein (white arrow). (b) Post-surgical CT angiography in transverse view revealing an abnormal pulmonary venous drainage into the right atrium (arrow).

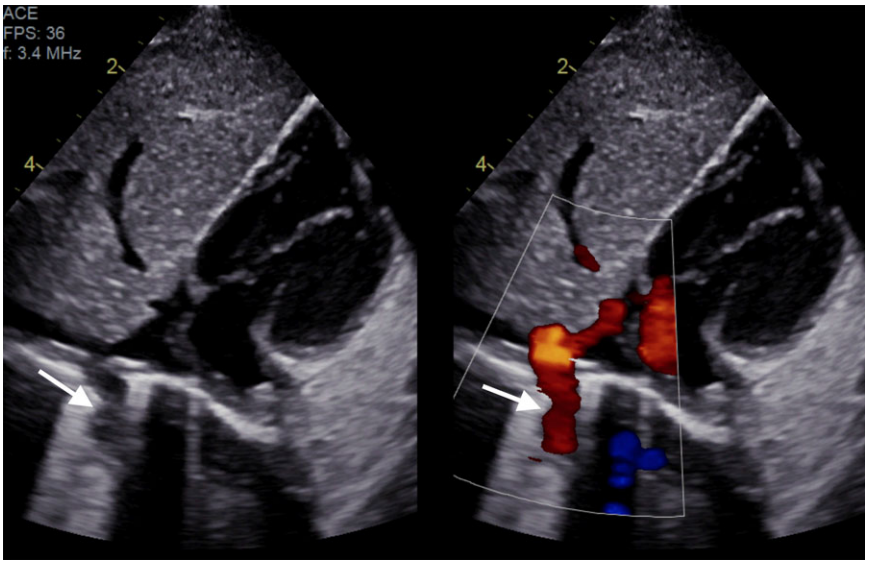

Figure 2. Transthoracic heart ultrasound revealing an anomalous drainage resulting from one vein originating in the lower portion of the right lung – scimitar vein (arrow) into the inferior vena cava.

Discussion

Despite unknown embryological development errors accounting for the anatomical features depicted in Scimitar syndrome, ranging from common cardiac and pulmonary abnormalities to less common vertebral and genitourinary tract anomalies, this entity has very rarely been described in association with diaphragm defects. To the best of our knowledge, only once has it been reported together with a case of diaphragmatic eventration.[3] Usually, two types of Scimitar syndrome occur: infantile and childhood/adult form. The former has a higher incidence and is usually associated with comorbidities that result from congestive heart failure, pulmonary hypertension, and other systemic congenital defects. Congenital cardiac defects were reported in 19–31% of Scimitar syndrome patients.[4] Diagnosis of this early age form usually occurs in the first month of life, presenting with failure to thrive, tachypnoea, and heart failure. Cyanosis may be present if there is pulmonary hypertension. Mortality is unfortunately still high soaring up to 45%.[4] In our case, much like *Clements* et al.[3] reported, respiratory symptoms presented very early, thus leading to quicker diagnosis and follow-up procedures. Conversely, the childhood/adult form of this syndrome presents with mild symptoms, if any, and is associated with low mortality rates. Child or adult patients usually have a higher chance of chest infections confined to the right lung and may experience heart dysrhythmias, due to right-sided volume overload.

Amongst several congenital anomalies that accompany the Scimitar syndrome, the horseshoe lung is known to be one of the most common with an incidence of 80–85% of cases.[5] The horseshoe lung configuration happens through the fusion of a common parietal pleural defect that provides communication between the pleural cavities. Based on anatomical features, a classification has been proposed with three distinct patterns: (1) the lungs show a complete fusion in baseline without intervening pleura; (2) a lung segment extends towards opposite hemithorax with a unilateral pleural layer; (3) bilateral pleural layers and horseshoe lung is a separate lobe in its own visceral pleura.[5] Our case is suggestive of a type 2 pattern since there was no evidence of pleural disruption. A recent case report has also reported a similar pattern.[5]

https://doi.org/10.1017/S1047951121001694 Published online by Cambridge University Press

Regarding clinical management, symptomatic DE cases are uncommon and sometimes difficult to distinguish from paralysis of the diaphragm. Surgical plication is recommended to promote a better quality of life.[2] In our case, it was not clear whether the DE or the Scimitar syndrome itself caused the onset of symptoms, but corrective surgery showed a good result. Outcomes in therapy on infantile Scimitar syndrome are suboptimal, due to significant associated congenital heart defects. On the other hand, patients with an isolated form of disease, presenting after infancy may have favourable outcomes with either medical or surgical approaches.[6] Since our case depicts an infantile form of disease, prognosis will need to be reassessed in time according to symptoms and the child's development.

In conclusion, Scimitar syndrome is a rare congenital disorder that may be associated with other congenital anomalies, but most commonly heart and pulmonary abnormalities. Symptoms present more frequently in the first month of life, but can be present early after birth. Diaphragmatic defects are only seldom reported in association with this disease and our case featured a DE as a primary finding in this syndrome.

Acknowledgements. The authors would like to acknowledge the collaboration of the Radiology Department of Hospital Santa Maria, namely Dr Luísa Lobo who acquired the images and guided the radiological discussion.

Financial support. The authors declare no funding sources in conducting this work.

Conflicts of interest. The authors declare no conflict of interest in conducting this work.

Ethical standards. The authors assert that all procedures contributing to this work comply with the ethical standards of the Helsinki Declaration of 1975, as revised in 2008. Consent for publication has been obtained.

References

1. Yang CK, Shih JC, Hsu WM, et al. Isolated right diaphragmatic eventration mimicking congenital heart disease in utero. Prenat Diagn 2005; 25: 872–875. doi: 10.1002/pd.1281.
2. Groth SS, Andrade RS Diaphragmatic Eventration. Thorac Surg Clin 2009; 19: 511–519. doi: 10.1016/j.thorsurg.2009.08.003.
3. Clements BS, Warner JO The crossover lung segment: congenital malformation associated with a variant of scimitar syndrome. Thorax 1987; 42: 417–419.
4. Çiçek S, Arslan AH, Ugurlucan M, Yildiz Y, Ay S Scimitar syndrome: The curved Turkish sabre. Semin Thorac Cardiovasc Surg Pediatr Card Surg Annu 2014; 17: 56–61. doi: 10.1053/j.pcsu.2014.01.003.
5. Gonen KA, Canitez Y, Bostan OM, Yazici Z Horseshoe lung associated with scimitar syndrome. BMJ Case Rep2019; 12: 10–13. doi: 10.1136/bcr-2014-204389.
6. Wang H, Kalfa D, Rosenbaum MS, et al. Scimitar Syndrome in Children and Adults: Natural History, Outcomes, and Risk Analysis. Ann Thorac Surg 2018; 105: 592–598. doi: 10.1016/j.athoracsur.2017.06.061.

https://doi.org/10.1017/S1047951121001694 Published online by Cambridge University Press