CASE REPORT

The Rare Congenital Anomaly of Scimitar Syndrome: An Unusual Presentation in Symptomatic Adult

Siti Zulaiha Binti Che Hat

Clinical Research Centre, Hospital Tuanku Fauziah, Perlis, Ministry of Health Malaysia

ABSTRACT

Scimitar syndrome is a rare congenital heart defect occurring in 1 to 3 per 100,000 live births. This is a case of a 26 years old lady presenting with episodic fainting spells since the age of 18 years old. She was initially diagnosed with epilepsy until a referral to our centre found a soft splitting of the second heart sound and multiple premature ventricular complexes on ECG. The computed tomography of the pulmonary artery confirmed the diagnosis if Scimitar syndrome in the presence of anomalous single right pulmonary vein draining into infra-diaphragmatic systemic venous circulation. A corrective open-heart surgery to re-implant the pulmonary vein was performed with excellent clinical outcomes. Therefore, it is crucial for clinicians to embody high index of suspicion of congenital anomaly even in adults presenting with indefinite clinical symptoms. This report also represents the first published case of adult Scimitar syndrome from Malaysia.

Keywords: Scimitar syndrome, Hypertension, pulmonary, Cardiomegaly, Heart defects

Corresponding Author:
Siti Zulaiha Che Hat, Diploma in Nursing
Email: sitizulaihacrc@gmail.com
Tel: +604-973 8000 (ext. 8413)

INTRODUCTION

Scimitar syndrome is exceedingly rare and occurs in only 1 to 3 per 100,000 live births. Described by George Cooper in 1836 while conducting an autopsy on a 10-month-old infant (1), the term "*Scimitar*" was first introduced by Halasz in 1956 (2) to describe a partial anomalous right pulmonary venous drainage into the inferior vena cava instead of into the left atrium.

Partial anomalous pulmonary venous return (PAPVR) is a rare congenital anomaly constituting only 0.5 to 1% of all congenital heart disease (2) characterised by the abnormal diversion of the drainage from the right side of the lung to the inferior vena cava (IVC) at the infra-diaphragmatic level or at the intersection between the IVC and the right atrium (2). Patients commonly present with respiratory symptoms in early infancy with entailed prompt surgical intervention, often with co-existence of associated anomalies such as the atrial septal defect (3). Treatment decisions however, are challenging in patients with isolated Scimitar syndrome, as they are often diagnosed by coincidence later during adolescence or later adulthood. These patients typically remain asymptomatic for several years and are able to lead a normal, non-debilitating life.

Diagnostic modality includes imaging with cardiac catheterisation as described by Dotter et al. in 1949 whilst the first surgical intervention was performed in 1950 by Drake and Lynch involving resection of the right lower lung (4). Over the years, the clinical management of the syndrome was improvised and the first corrective surgery was done in 1956 by Schramel et al (4). They have recognised two variations of scimitar veins; a simple classic vein that goes from the middle portion of the right lung running to the cardiophrenic angle and a second type, in which a double-arched vein in the lower and upper zones of the lungs draining concurrently into the left atrium and inferior vena cava.

Scimitar syndrome is regularly diagnosed during infancy, therefore findings in adult cases are extremely rare. Adult cases of Scimitar syndrome tend to be presenting with only mild symptoms and often carry a benign prognosis. However, this is a complicated case of an adult form Scimitar syndrome with a rare presentation that merits clinical recognition, hence is reported.

CASE REPORT

A 26 years old lady presented to our outpatient department with a history of syncopal attack during hiking 4 days prior. The syncopal attack was preceded by palpitation, shortness of breath and light-headedness. There was also a presence of aura described as "green light" flashes before she passed out and regained consciousness spontaneously after 10 to 15 seconds. She was seen to develop an up-rolling of the eyeballs during the black-out episode.

Upon further questioning, the patient has had multiple episodes of syncopal attack and was admitted several

times since the age of 18 years old. She was initially diagnosed with epilepsy, however no anti-epileptic medication was started in view of the irregular interval. The electroencephalogram (EEG) and the computed tomography (CT) of the brain were reported to be normal. The episodic fainting spells typically occur when she was under physical or emotional stress and they were typically brief with rapid recovery. Apart from that, she has had several history of recurrent pneumonia requiring antibiotics over the years. The infection typically responds well to treatment with no long-term follow-up given. She also reported of irregular palpitation episodes and dyspnoea, particularly on strenuous exertion. There was no history of chest discomfort or swelling in her ankles or legs. Otherwise, there was no known other medical illness. Her parents were non-consanguineous. There was no family history of congenital anomalies or syndromic children in the family. Her birth history was uneventful.

On examination, her vital signs were stable (Blood pressure: 105/67 mmHg, Pulse rate:89 bpm, Afebrile). Examination revealed a soft splitting of the second heart sound, best heard over the pulmonic region. There were no thrills or parasternal heave. Her baseline routine laboratory results were within normal limits (Potassium: 4.2 mmol/L, Urea: 4.2 mmol/L, Creatinine: 57µmol/L, Hb: 14.3 g/dL). Thyroid function test was also within normal range.

Electrocardiogram (ECG) done showed multiple premature ventricular complexes (PVC) (Fig. 1) and right bundle branch block (Fig. 2). Her chest x-ray (CXR) showed enlargement of the right heart (Fig. 3) while the ECHO bubble test was negative. Holter test revealed presence of supraventricular beats and the right heart study with contrast revealed an enlarged right ventricle and dilated pulmonary artery with pulmonary artery pressures of 8 to 20 mmHg with a mean of 15 mmHg. Additionally, computed tomography of the pulmonary artery (CTPA) with contrast revealed the diagnosis of partial anomalous pulmonary venous return of the right pulmonary veins, uncovering the inferior vena cava being located at the right atrial junction (Fig. 4a), draining into the intrahepatic IVC (Fig. 4b) and was complicated with pulmonary hypertension, mild cardiomegaly and minimal atelectasis over her right middle lobe. The main pulmonary artery appeared dilated with the antero-posterior (AP) diameter of 3.14 cm at its bifurcation level.

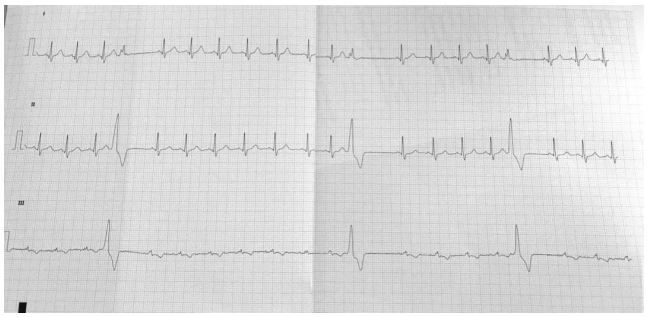

Figure 1: Electrocardiogram (ECG) showing multiple PVCs

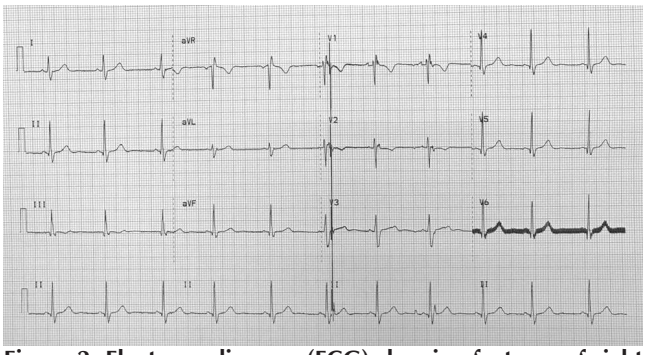

Figure 2: Electrocardiogram (ECG) showing features of right bundle branch block

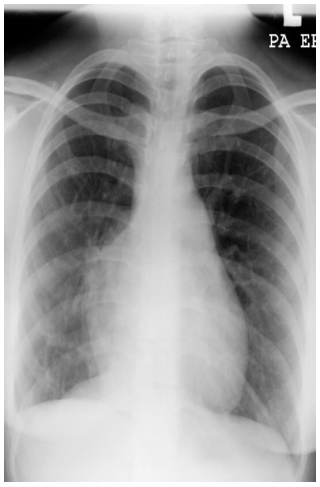

Figure 3: CXR showing hypertrophy of the right side of the heart

Upon confirmation of diagnosis, the patient was referred to a tertiary cardiac centre and an open corrective heart surgery involving re-implantation of pulmonary vein requiring cardiopulmonary bypass with deep hypothermic circulatory arrest was performed. Post-operative Day-3, she developed mild pleural effusion requiring oral Lasix 40 mg tds. Incentive spirometry was also encouraged to prevent atelectasis. Patient was discharged well on Day-8 hospitalisation with complete resolution of pleural effusion. The patient is currently on cardiac rehabilitation program and was compliant to oral Bisoprolol 2.5 mg daily to control her pulmonary hypertension. She is otherwise well, able to perform her daily activities without physical limitation and did not experience further episode of syncopal attack to date.

DISCUSSION

We report a missed case of Scimitar syndrome in a young lady that first manifests in her adolescent years. Scimitar syndrome is a rare congenital disorder with varied clinical presentation. Hence, the diagnosis can be easily missed due to its mild to moderate course of clinical symptoms. The infantile form of Scimitar syndrome presents with signs of respiratory distress in

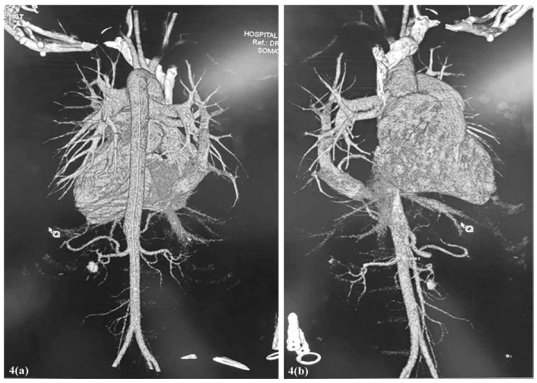

Figure 4: 3D-reconstructive images from computed tomography angiographic scan of the pulmonary vessels confirming Scimitar syndrome

early infancy period whereby prompt surgical correction is the conventional mode of treatment (1). On the other hand, the adult form of Scimitar syndrome is typically mild and carries a benign course without the need of surgical intervention. However, complicated cases may present with recurrent pneumonia, particularly on the right side with signs of pulmonary hypertension and chronic right ventricular overload (5) that may benefit from surgical treatment (3).

Our case represents the adult form of complicated Scimitar syndrome, presenting with episodic syncopal attack and recurrent pneumonia. Scimitar syndrome may cause a significant left to right shunt from the anomalous pulmonary venous drainage at the atrial level which contributes to the right ventricular volume overload (4)(5) and increases the risk of pulmonary infection. Due to the rarity of this syndrome, there is no well-defined mechanism by which recurrent pneumonia in cases of Scimitar syndrome may develop. However, it was suggested that recurrent chest infection may result from the extrinsic compression of the airway from the Scimitar vein (5), or contributed by the impaired venous drainage (4).

Complicated cases of Scimitar syndrome that progresses without any intervention may be associated with severe pulmonary hypertension that may lead to premature death. Therefore, early diagnosis and monitoring can improve prognosis and the patients' quality of life. Our patient presented with episodic fainting spells since her adolescent years and her symptoms were initially dismissed as mere epilepsy. Her illness was already complicated with pulmonary hypertension at the time the diagnosis of Scimitar syndrome was made. In this case, the chest x-ray may provide a clue in the diagnosis of Scimitar syndrome by the presence of a scimitar-shaped shadow created by the curvilinear density along the right heart border by the descending vein (2)(4)(5). Electrocardiography typically showed features of right ventricular hypertrophy in 50% of Scimitar syndrome and features of right bundle branch block in 10% of patients (4,5).

A complicated case of adult form Scimitar syndrome is exceedingly rare that we only found very few similar cases from our extensive literature search. Surgical management is recommended in the presence of pulmonary hypertension with right-sided heart failure (1,3,5), as seen in our case. Additionally, Scimitar syndrome management plan requires an inter-professional team approach that includes a paediatrician, paediatric cardiologist, cardiac surgeon, and a radiologist. A missed case of Scimitar syndrome may lead to poor clinical outcomes as the disease may progress with irreversible complications. Hence, a high index of suspicion coupled with an advanced imaging resources will facilitate surgical strategy, resulting in reduced morbidity and mortality (1,2).

CONCLUSION

Our case highlights the rare presentation of an adult case of Scimitar syndrome who presented with recurrent syncopal attack and recurrent chest infections that were earlier dismissed as isolated case of epilepsy and community-acquired pneumonia. We have also successfully underscore the importance of developing a high index of suspicion of a congenital anomaly even in an adult presenting with vague clinical symptoms with inconclusive diagnosis.

AKNOWLEDGEMENTS

The author would like to thank Dr Karniza Khalid from Clinical Research Centre, Hospital Tuanku Fauziah, Perlis for sharing her pearls of wisdom in proofreading this case report. The author would also like to thank Dr Othman bin Warijo, the Director of Hospital Tuanku Fauziah, Perlis for his continuing support of institutional research endeavours. The study was registered with the National Medical Research Registry of Ministry of Health Malaysia with the registration no. NMRR-19-3676-52111.

REFERENCES

1. Wang H, Kalfa D, Rosenbaum MS, Ginns JN, Lewis MJ, Glickstein JS, et al. Scimitar Syndrome in Children and Adults: Natural History, Outcomes, and Risk Analysis. Ann Thorac Surg. 2018 Feb;105(2):592-598.
2. Holt PD, Berdon WE, Marans Z, et al. Scimitar vein draining to the left atrium and a historical review of the scimitar syndrome. Pediatr Radiol. 2004;34(5):409-413.
3. Vida VL, Padalino MA, Boccuzzo G, Tarja E, Berggren H, Carrel T, et al. Scimitar syndrome: A European congenital heart surgeons association (ECHSA) multicentric study. Circulation. 2010; 122(12): 1159-1166.
4. Зiзek S, Arslan AH, Ugurlucan M, Yildiz Y, Ay S. Scimitar syndrome: the curved Turkish sabre. In Seminars in Thoracic and Cardiovascular Surgery: Pediatric Cardiac Surgery Annual 2014 Jan 1 (Vol. 17, No. 1, pp. 56-61). WB Saunders.
5. Jackson N, Nokes BT, Sakata K, Cummings K, Vaszar L. An unusual variant of scimitar syndrome predisposing to recurrent pneumonia. Resp Med Case Rep. 2019 Jan 1;26:240-3.