CRCP Case Reports in Clinical Practice

Case Report

Journal Homepage: http://crcp.tums.ac.ir

A Rare Case of Partial Anomalous Pulmonary Venous Return (Scimitar Syndrome) with Vaginal Agenesis and a History of Infantile Imperforated Anus

Maziar Karamnejad[1], Mahdis Nazari[2*], Ehsan Parvas[1], Reza Elahi[3], Amir Ghaffari Jolfayi[4], Kyomars Abbasi[1]

1. Research Center for Advanced Technologies in Cardiovascular Medicine, Tehran Heart Center, Tehran University of Medical Sciences, Tehran, Iran.

2. Zanjan Metabolic Disease Research Center, Health and Metabolic Disease Research Institute, Zanjan University of Medical Sciences, Zanjan, Iran.

3. Department of Radiology, Zanjan University of Medical Sciences, Zanjan, Iran.

4. Cardiovascular Research Center, Rajaie Cardiovascular, Medical, and Research Center, Iran University of Medical Sciences, Tehran, Iran.

Use your device to scan and read the article online

Citation Karamnejad M, Nazari M, Parvas E, Elahi R, Ghaffari Jolfayi A, Abbasi K. A Rare Case of Partial Anomalous Pulmonary Venous Return (Scimitar Syndrome) with Vaginal Agenesis and a History of Infantile Imperforated Anus. Case Reports in Clinical Practice. 2025; 10(1): 7-11. DOI:10.18502/crcp.v10i1.19473

Running Title Partial Anomalous Pulmonary Venous Return with Vaginal Agenesis

Article info:
Received: December 28, 2024
Revised: January 25, 2025
Accepted: February 22, 2025

Keywords:
Partial anomalous pulmonary venous return (PAPVR); Scimitar syndrome; Imperforate anus; VACTERL association; Vaginal agenesis

ABSTRACT

Scimitar syndrome is a rare congenital cardiac anomaly characterized by abnormal drainage of the right pulmonary veins into the inferior vena cava (IVC). Here, we report the case of a 26-year-old female diagnosed with scimitar syndrome (SS), with a history of imperforate anus and vaginal agenesis, consistent with a VACTERL association. The imperforate anus was identified and treated in infancy, whereas the vaginal agenesis was diagnosed at menarche. Additionally, the patient presented with a large atrial septal defect (ASD) and a hypoplastic right lung alongside SS. This case underscores the potential correlation between SS, vaginal agenesis, and imperforate anus as features within the VACTERL spectrum.

Introduction

Scimitar syndrome (SS) is a rare congenital condition classified under partial anomalous pulmonary venous return (PAPVR) [1]. It is characterized by a left-to-right shunt at the atrial level and is associated with various anatomical anomalies, including right lung hypoplasia, abnormal pulmonary venous drainage, atypical arterial supply to the lung lobes, bronchial tree malformations, and multiple cardiac defects. Among these, atrial septal defect (ASD) is the most frequently associated cardiac anomaly; however, other conditions such as patent ductus arteriosus (PDA), ventricular septal defect (VSD), and tetralogy of Fallot (TOF) may also occur [2].

A unique feature of SS is the potential dextroposition

*** Corresponding Author:**
Mahdis Nazari
Address: *Zanjan Metabolic Disease Research Center, Health and Metabolic Disease Research Institute, Zanjan University of Medical Sciences, Zanjan, Iran*
E-mail: *mahdisnazari96@gmail.com*

of the heart, which further complicates its presentation. The clinical manifestations of scimitar syndrome are highly variable, reflecting the diversity in anatomical variations and the extent of left-to-right shunting. While some individuals—particularly adults and older children—may remain asymptomatic, infants can experience severe complications such as cardiopulmonary distress, pulmonary hypertension, and heart failure, which may be life-threatening within the first year of life. Scimitar syndrome is often identified incidentally in childhood or later in life when secondary symptoms appear, highlighting the importance of thorough evaluation [3].

The term *scimitar syndrome* originates from the distinctive tubular opacity seen on chest radiographs, which follows the right cardiac border and resembles the shape of a traditional scimitar sword. Advanced imaging modalities, including computed tomography angiography (CTA) and cardiac magnetic resonance (CMR) imaging, are essential for confirming the diagnosis, with echocardiography aiding in the identification of associated cardiac anomalies [3].

Scimitar syndrome can sometimes co-occur with the VACTERL association, a rare grouping of congenital anomalies that includes vertebral defects, anal atresia, cardiac abnormalities, tracheoesophageal fistula, renal or genitourinary anomalies, and limb deformities [4]. Of particular concern within this association is the uncommon combination of anorectal anomalies with vaginal agenesis, which may go undiagnosed until menarche, potentially leading to significant medical complications. Effective management of Scimitar syndrome requires a multidisciplinary approach, integrating medical treatment for heart failure symptoms with surgical correction of anatomical abnormalities.

Case presentation

A 26-year-old woman was evaluated for recurrent pneumonia, exertional respiratory distress, and unexplained fatigue. She had no fever or other cardiopulmonary symptoms but had a history of congenital anovaginal defects corrected surgically in infancy and adolescence, including imperforate anus repair and vaginal reconstruction for agenesis. She also had a childhood repair of an atrial septal defect (ASD) and vertebral surgery following a fracture (Figure 1). Electrocardiographic findings were unremarkable and consistent with a hemodynamically stable state (Figure 2).

Physical examination revealed a stable patient with a systolic murmur at the lower right sternal border, likely related to anomalous venous flow. Chest X-ray showed an abnormal curved structure near the right lung. Electrocardiography was normal, and echocardiography demonstrated mild right ventricular enlargement, mild tricuspid and mitral regurgitation, and an estimated systolic pulmonary artery pressure of 28 mmHg. Findings were suggestive of partial anomalous pulmonary venous connection (PAPVC), raising suspicion for Scimitar syndrome. Unfortunately, the echocardiographic (TDE) views

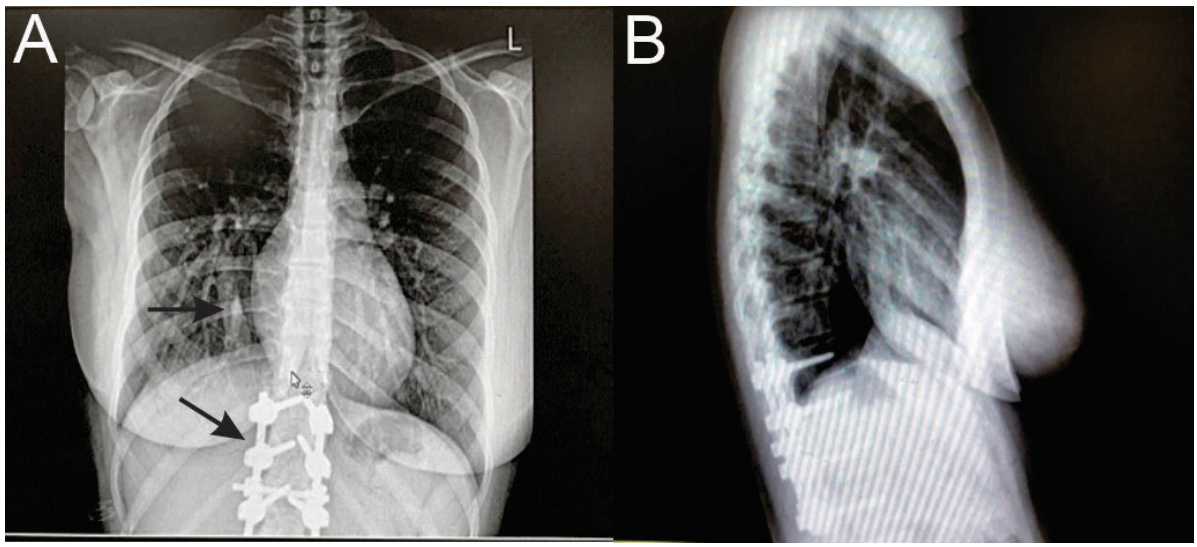

Fig. 1. Chest x-ray showing a scimitar-like abnormal shadow in the right lower lung field (arrows). The lower arrow demonstrates the history of previous thoracic spinal injury.

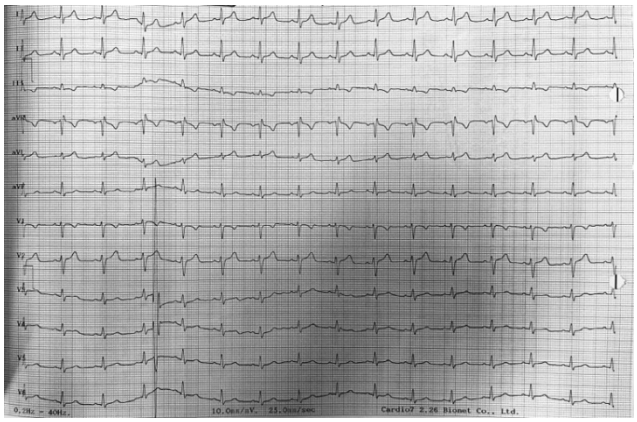

Fig. 2. A standard 12-lead electrocardiogram (ECG) from the patient.

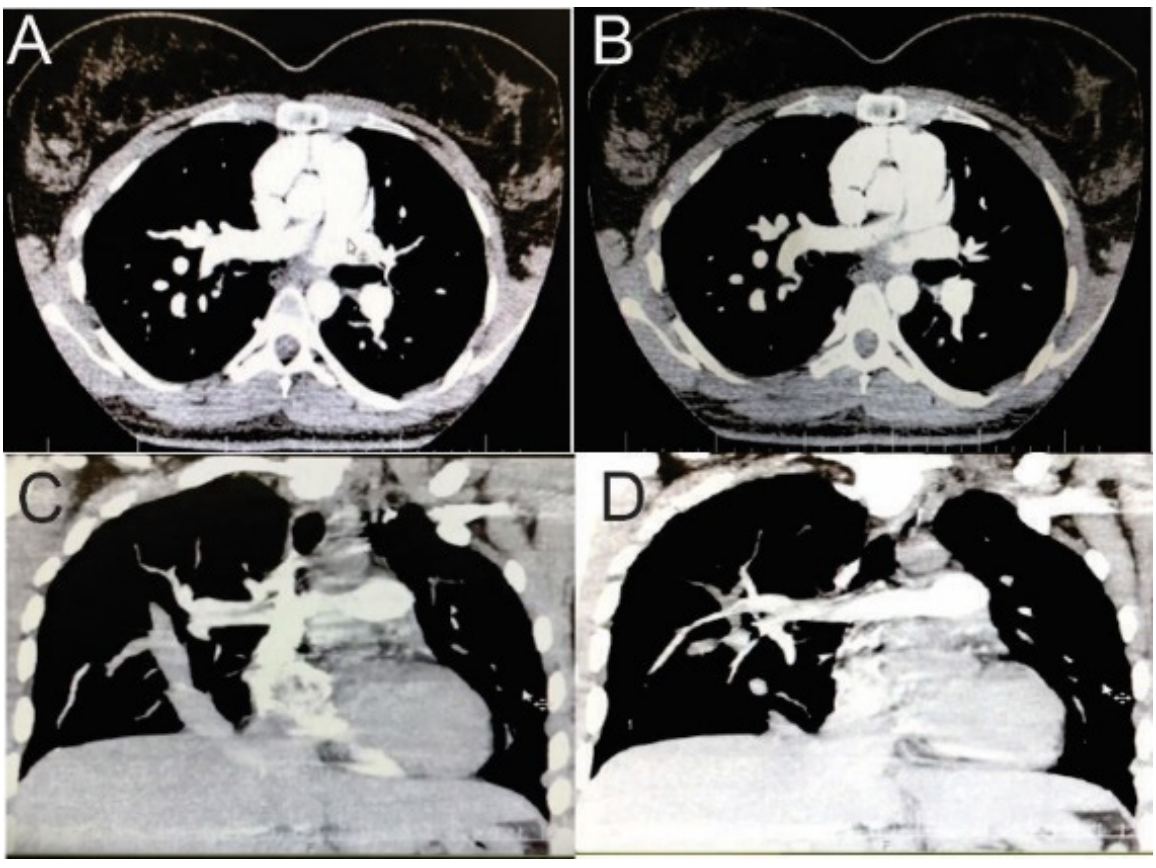

Fig. 3. A) A transverse section of a CTPA shows that a scimitar vein emerges from the right lung, draining the lower right lung into the inferior vena cava (IVC) at its junction with the right atrium. The right ventricle also appeared mildly enlarged. B) The coronal view of CTPA clearly illustrates a dilated, directing anomalous pulmonary vein in the right lower lung that drains into the IVC.

were not available, which is a limitation of our study.

Cardiac CT angiography (Figure 3) confirmed anomalous drainage of the right lower pulmonary veins into the inferior vena cava (IVC), with a left-to-right shunt (Qp/Qs ratio of 1.54), mild right ventricular dilation, and normal intracardiac pressures. Right heart catheterization confirmed normal chamber pressures and an oxygen saturation step-up consistent with a left-to-right shunt at the pulmonary venous level, confirming the diagnosis of Scimitar syndrome.

The patient's congenital anomalies were managed surgically: imperforate anus repair in infancy, vaginal reconstruction at 15, and ASD repair in childhood. Due to symptomatic right ventricular enlargement

and shunting, she underwent surgery to reroute the anomalous pulmonary vein from the IVC to the left atrium, restoring normal pulmonary venous return and reducing right heart overload. Surgical techniques are individualized; in this case, intra-atrial rerouting was performed.

Discussion

Scimitar syndrome is a rare congenital disorder identified by partial anomalous pulmonary venous return, which may be associated with various anatomical anomalies. Possible long-term complications from an atrial-level shunt include atrial arrhythmias from right atrial dilation, right ventricular dysfunction due to volume overload, and pulmonary hypertension from increased pulmonary flow, causing chronic clinical symptoms [5]. Patients diagnosed with Scimitar syndrome typically undergo comprehensive evaluations that include CT scans and cardiac angiography to accurately delineate the anomalous vessels associated with the condition. Recent recommendations have included cardiac MRI as an effective tool for assessing congenital abnormalities, further enhancing diagnostic capabilities in such cases. Our clinical presentation combined chest radiography, contrast-enhanced CTPA, and echocardiograms. Cases similar to ours, though not all, may necessitate more invasive procedures such as cardiac catheterization to evaluate the blood vessels. This multifaceted approach provided sufficient diagnostic information to identify Scimitar syndrome while ruling out other potential anomalies. As mentioned before, the majority of individuals affected by Scimitar syndrome are symptomatic infants, with limited documentation of adult cases. Moreover, adult patients with delayed diagnosis often have a normal lifespan without undergoing surgical intervention. Due to the symptoms and RV enlargement in our patient, both the cardiac surgery consultant and the patient agreed to proceed with corrective surgery.

Previously documented surgical techniques for managing Scimitar syndrome include direct anastomosis of the scimitar vein to the left atrium, intra-atrial rerouting after anastomosing the scimitar vein to the right atrium, and intra-IVC baffle rerouting [6]. A newer approach involves graft anastomosis of the scimitar vein to the right atrium and intra-atrial rerouting using autologous pericardium. Although this technique may result in longer hospital stays and necessitate anticoagulant therapy, it is recommended for cases of total anomalous right pulmonary venous drainage, where the left atrium is often underdeveloped [7]. For example, in one case, the scimitar vein drained into the distal inferior vena cava, which increased the distance to the left atrium. An alternative method employs a ringed Gore-Tex graft for anastomosis to the right atrium, combined with intra-atrial rerouting. This approach has generally yielded favorable outcomes, demonstrating low post-surgical pulmonary vein obstruction rates during a follow-up period of 3.6 years [7].

Missed diagnosis of PAPVC during surgical repair of ASD has been reported in the literature. Perri et al[8] . reported a 20-year-old male who developed recurrent pleural effusion and right heart failure symptoms years after surgical repair of a secundum ASD. Further imaging revealed previously undiagnosed PAPVC, with two pulmonary veins draining into the superior vena cava (SVC). The patient underwent a modified Warden procedure using a prosthetic conduit to redirect the anomalous pulmonary venous flow into the left atrium. The surgery was successful, with the patient recovering well, and follow-up imaging confirmed good surgical results. The authors highlighted the importance of thorough preoperative assessment for PAPVC in ASD patients, especially when unexplained right heart symptoms persist after ASD repair. They recommended the modified Warden procedure with a conduit as an effective surgical option for adult reintervention in cases of high SVC connection.

In another study in 2022, Perchik et al[9]. described a 33-year-old woman with a history of PAPVR and a superior sinus venosus defect who developed SVC syndrome three years after undergoing the Warden procedure. She presented with acute headache and facial swelling, and imaging revealed SVC anastomotic narrowing with thrombosis and extensive collateral venous drainage. Emergent endovascular intervention, including thrombectomy, angioplasty, stent placement in the SVC, and occlusion of the accessory hemiazygos vein, successfully resolved her symptoms and restored normal venous flow. The report highlights that while the Warden procedure is generally well-tolerated, delayed SVC obstruction can occur in adults, leading to right-to-left shunting through venous collaterals. Prompt recognition and minimally invasive management are crucial for favorable outcomes, and new venous varices in post-Warden patients should raise suspicion for evolving SVC stenosis.

Several studies suggest that anomalies in the vertebral, anal, and genitourinary systems may co-occur due to shared embryonic origins [10]. Genital and urinary anomalies are more common in VACTERL patients with anorectal atresia and renal defects, but less so in those with limb or esophageal anomalies. The genetic relationship between Scimitar syndrome

and vaginorectal defects remains unclear due to limited genetic testing [11].

Conclusions

Further research is needed to explore potential common pathogenic mechanisms linking Scimitar syndrome, anovaginal malformations, and other VACTERL-associated anomalies. Clinicians are advised to maintain high vigilance for cardiac defects, especially Scimitar syndrome, in neonates presenting with imperforate anus or genitourinary anomalies.

Ethical Considerations

Ethics approval and consent to participation

The patient gave informed consent. The patient was informed about the nature of the report, its purpose, and the potential implications of sharing the patient's medical information. They were assured that all identifying details would be omitted and the patient's privacy would be protected.

Funding

No funding was received to assist with the preparation of this manuscript.

Conflict of Interests

The authors have no conflict of interest to declare.

References

[1] Mounir R, Nya F, Mohammed B, Ayad A, Bamous M. Adults forms of scimitar syndrome. J Card Surg. 2020;35(7):1697-9. https://doi.org/10.1111/jocs.14625

[2] Bo I, Carvalho JS, Cheasty E, Rubens M, Rigby ML. Variants of the scimitar syndrome. Cardiol Young. 2016;26(5):941-7. https://doi.org/10.1017/S1047951115001651

[3] Aristizabal AM, Guzmán-Serrano CA, Mondol-Villamil NV, Bolaños-Vallejo LM, Mejia-Quiñones V, Recio-Gómez MA, et al. Clinical characteristics, imaging findings, management, and outcomes of patients with scimitar syndrome at a tertiary referral healthcare center in Colombia. Int J Cardiovasc Imaging. 2024;40(6):1319-28. https://doi.org/10.1007/s10554-024-03102-1

[4] Fritz CJ, Reutter HM, Herberg U. Scimitar syndrome in a case with VACTERL association. Cardiol Young. 2015;25(3):606-9. https://doi.org/10.1017/S1047951114000924

[5] Wang CC, Wu ET, Chen SJ, Lu F, Huang SC, Wang JK, et al. Scimitar syndrome: incidence, treatment, and prognosis. Eur J Pediatr. 2008;167(2):155-60. https://doi.org/10.1007/s00431-007-0441-z

[6] Seymour E, Mallory G, Morales-Demori R. Surgical and cardiac catheterization outcomes of scimitar syndrome patients: a three decade single-center experience. Pediatr Cardiol. 2023;44(3):579-86. https://doi.org/10.1007/s00246-022-02965-2

[7] Geggel RL, Gauvreau K, Callahan R, Feins EN, Baird CW. Scimitar syndrome: a new multipatch technique and incidence of postoperative pulmonary vein obstruction. JTCVS Tech. 2020;4:208-16. https://doi.org/10.1016/j.xjtc.2020.07.027

[8] Perri G, Graziani F, Bruno P, Grandinetti M, Lanzillo C, Marziali M, et al. Modified Warden procedure in adult with partial anomalous pulmonary venous connection after previous atrial septal defect repair. Cor Vasa. 2016;58(5):e501-4. https://doi.org/10.1016/j.crvasa.2015.08.004

[9] Perchik JD, Wilson CM, Abozeed M, Manapragada PP, Ahmed AN, Singh SP. Adult partial anomalous pulmonary venous return repair and superior vena cava syndrome: a delayed complication of the Warden procedure. Radiol Cardiothorac Imaging. 2022;4(5):e220077. https://doi.org/10.1148/ryct.220077

[10] Solomon BD, Raam MS, Pineda-Alvarez DE. Analysis of genitourinary anomalies in patients with VACTERL (vertebral anomalies, anal atresia, cardiac malformations, tracheo-esophageal fistula, renal anomalies, limb abnormalities) association. Congenit Anom (Kyoto). 2011;51(2):87-91. https://doi.org/10.1111/j.1741-4520.2010.00303.x

[11] Botto LD, Khoury MJ, Mastroiacovo P, Castilla EE, Moore CA, Skjaerven R, et al. The spectrum of congenital anomalies of the VATER association: an international study. Am J Med Genet. 1997;71(1):8-15. https://doi.org/10.1002/(SICI)1096-8628(19970711)71:1<8::AID-AJMG2>3.0.CO;2-V