The Sword and the Crown: Echocardiography for the Detection of a Rare Combination of Congenital Heart Disease

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INTRODUCTION

The echocardiographic evaluation of shunts in adults is often challenging due to their larger body size, which may impair visualization of cardiac structures. Close inspection of all transthoracic echocardiography (TTE) images is critical to identify subtle clues that may indicate the presence of a shunt, particularly in patients with unexplained cardiac chamber remodeling. Accordingly, we present a young patient with unexplained right ventricular enlargement who was found to have partial anomalous pulmonary venous return (PAPVR), which was detected by TTE. We will review the epidemiology, natural history, cardiac imaging findings, and management of this rare condition.

CASE PRESENTATION

A 35-year-old woman with a medical history of peripheral neuropathy, dextroposition, and newly diagnosed left breast invasive ductal carcinoma presented to the hospital with 1 day of substernal chest pain. The chest pain began suddenly, worsened on exertion, and radiated to their bilateral subcostal chest. The physical exam was unremarkable for cardiopulmonary findings but demonstrated bilateral lower extremity weakness consistent with known polyaxonal neuropathy. A previous TTE obtained 8 years prior showed dextroposition with right-sided chamber enlargement, which was not further evaluated. Their presenting electrocardiogram showed sinus tachycardia to a heart rate of 110 with no ischemic findings. Serial serum troponins were normal at <6 ng/L and 10 ng/L on arrival and 3 hours after, respectively. A TTE was ordered for further assessment of patient's chest pain.

A TTE demonstrated right ventricle (RV) dilation with normal systolic function (Figure 1A, Video 1); RV end-diastolic diameter, tricuspid annulus planar excursion, and RV S′ were 48 mm, 17 mm, and 19 cm/sec, respectively. Left ventricular ejection fraction was normal at 56% (biplane method of disks) without regional wall motion abnormalities. The right atrium was mildly enlarged (indexed volume, 34 mL/m^2). Peak tricuspid velocity (2.2 m/sec) and estimated RV systolic pressure (22 mm Hg) were normal. On subcostal view, abnormal flow was visualized entering the inferior vena cava (IVC) posteriorly (Figure 1B, Video 2). The coronary sinus (CS) was severely dilated (22 mm); on agitated saline study performed from a left-sided intravenous cannula, microcavitations were visualized in the CS prior to entering the RV, suggestive of a persistent left-sided vena cava (PLSVC; Figure 1C–E, Video 3). Given the findings seen on TTE, a chest computed tomography (CT) scan with contrast was obtained for further assessment of mediastinal structures, vasculature, and lung fields. Imaging revealed PAPVR with both right-sided veins draining inferiorly into the right-sided IVC (Figure 2, Videos 4 and 5), findings consistent with scimitar syndrome. A PLSVC was also noted, confirming the findings seen on TTE. Multiple small, left subsegmental pulmonary emboli were also seen. Although not previously reported, the

VIDEO HIGHLIGHTS

Video 1: Two-dimensional TTE, RV-focused apical 4-chamber view, demonstrates the right heart dilation with normal RV function.

Video 2: Two-dimensional TTE, subcostal view with color-flow Doppler, demonstrates abnormal blood flow into the IVC posteriorly, which was ultimately found to be due to PAPVR with scimitar syndrome.

Video 3: Two-dimensional TTE, parasternal long-axis view during left arm injection of agitated saline contrast, demonstrates the CS dilation with entry of microcavitations first seen in the CS and subsequently in the RV consistent with the PLSVC.

Video 4: Contrast-enhanced chest CT, axial stack, superior to inferior display, demonstrates the contrast entering via the left arm and the PLSVC to the dilated CV. The anomalous right pulmonary vein is seen draining into the IVC (PAVPR), consistent with scimitar syndrome.

Video 5: Three-dimensional whole-heart CT, volume-rendered rotational display, demonstrates the pulmonary venous drainage (*blue*) with right-sided PAPVR and SVC drainage (*red*) indicating PLSVC.

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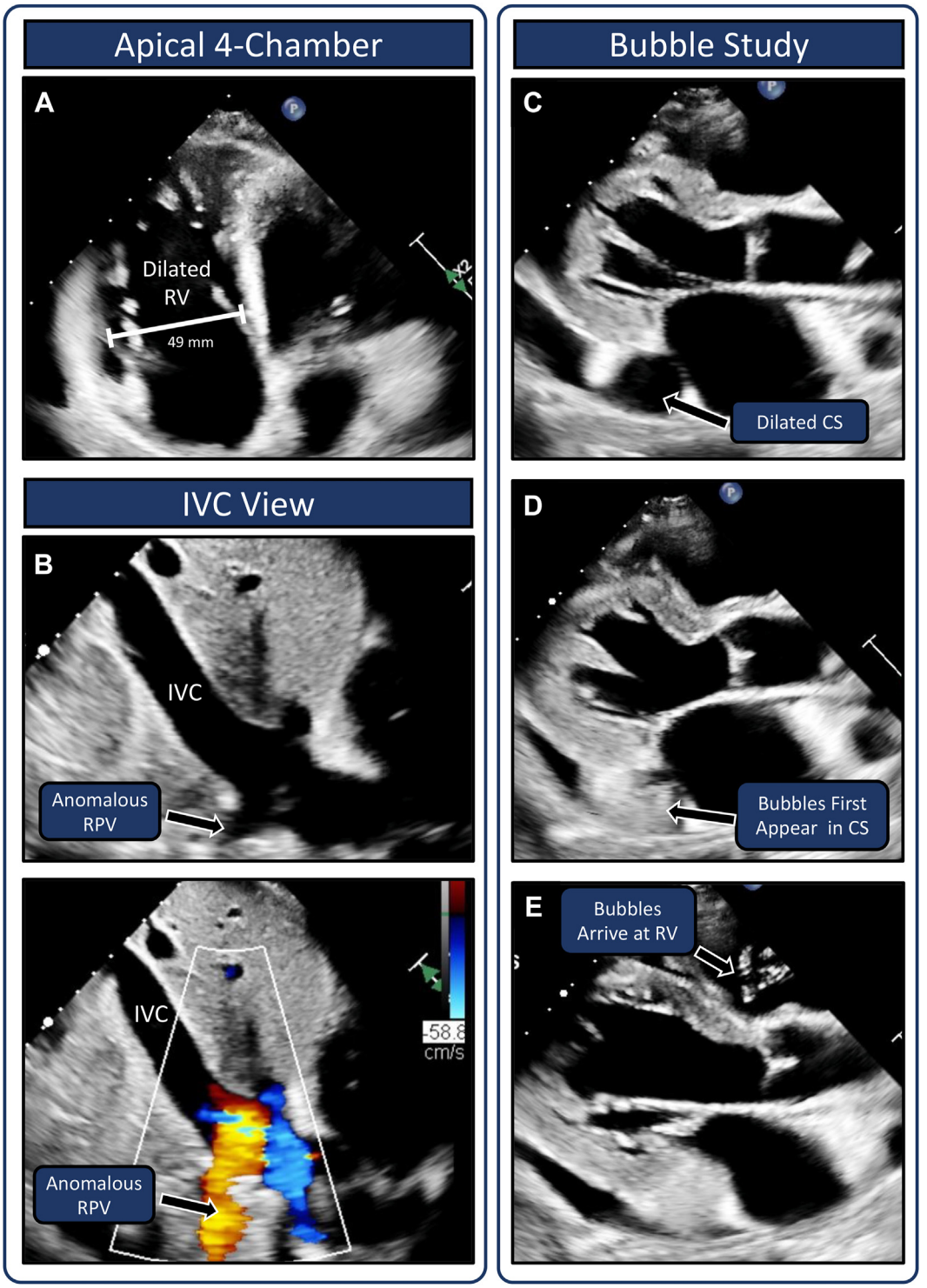

Figure 1 **(A)** Two-dimensional TTE, apical 4-chamber view, end-diastolic phase, demonstrates the dilated RV. **(B)** Two-dimensional TTE, subcostal view without (*top*) and with (*bottom*) color-flow Doppler, demonstrates abnormal blood flow into the IVC posteriorly, which was ultimately found to be due to PAPVR with scimitar syndrome. Two-dimensional TTE, sequential parasternal long-axis views obtained after left arm injection of agitated saline contrast, demonstrates the CS dilation **(C)** with entry of microcavitations first seen in the CS **(D)** and subsequently in the RV **(E)** consistent with the PLSVC. *RPV*, Right pulmonary vein.

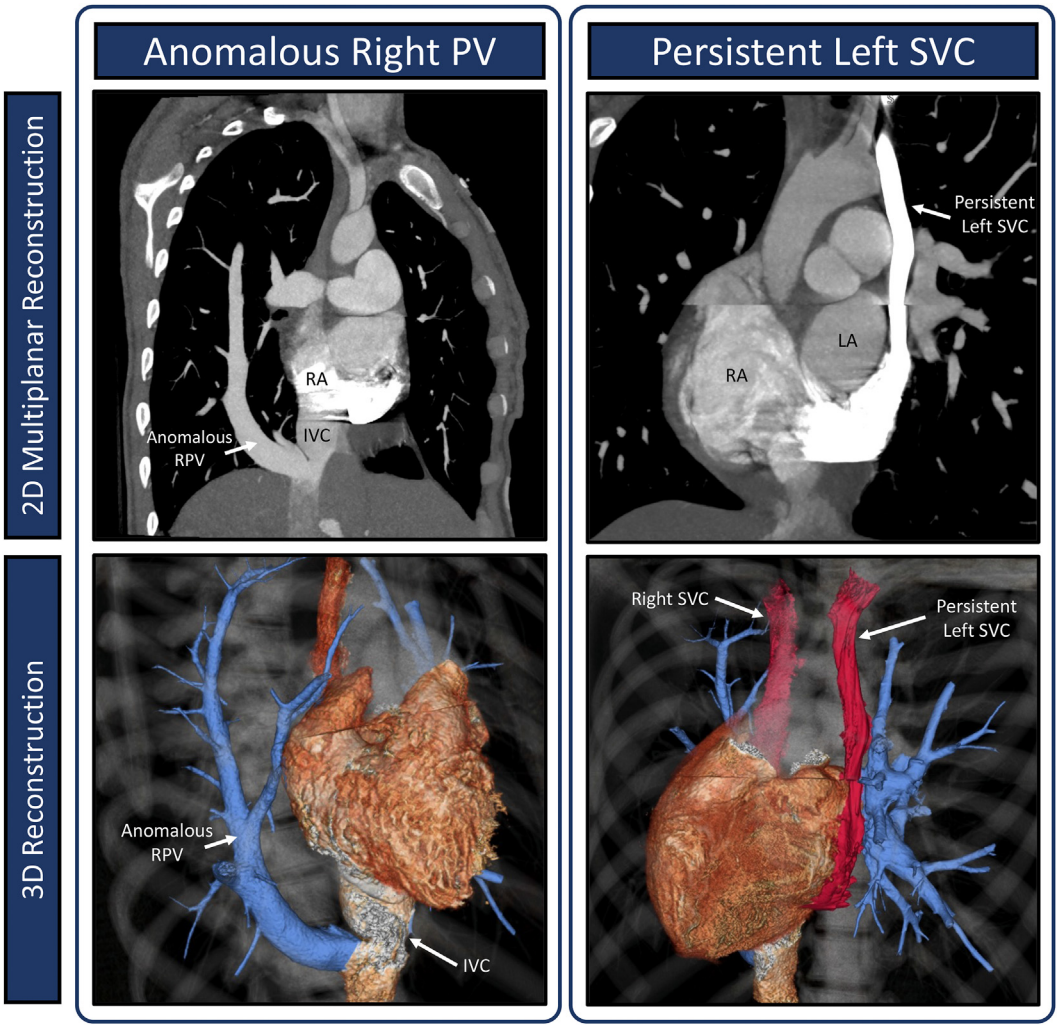

Figure 2 Multiplanar CT reconstructions (*top row*) and color-coded, volume-rendered three-dimensional (3D) whole-heart reconstructions (*bottom row*) demonstrate the PAPVR with drainage of the right-sided pulmonary veins (RPVs) into the IVC (*left panels*), consistent with scimitar syndrome. The *right panels* confirm the presence of the PLSVC draining into the dilated CS. *2D*, Two dimensional.

anomalous right pulmonary vein was able to be visualized on close inspection of the presenting chest x-ray (Figure 3).

While in the hospital, the patient was started on rivaroxaban. They are being followed by oncology and will begin neoadjuvant chemotherapy with cyclophosphamide and adriamycin and subsequent left breast lumpectomy. They were evaluated by the adult congenital heart disease team with plans to complete a right heart catheterization to confirm the absence of pulmonary hypertension and quantify the degree of right-to-left shunting. This will help decide candidacy for surgery, which is indicated in those with significant shunting—defined as Qp/Qs >1.5:1—assuming pulmonary artery pressures and pulmonary vascular resistance are ≤1/2 and 1/3 of systemic values, respectively.[1] Given the active malignancy and relative lack of symptoms attributable to the PAPVR, surgery would ideally be deferred pending remission from cancer. If the patient is unable to achieve remission and develops symptoms refractory to medical management, this would require a complex multidisciplinary discussion and patient-centered approach to determine the best treatment option.

DISCUSSION

Unexplained right heart dilation on TTE in an adult patient should raise suspicion for a left-to-right shunt. Pretricuspid shunts such as atrial septal defect (ASD), unroofed CS, or PAPVR lead to right heart dilation, while posttricuspid shunts such as ventricular septal defects and patent ductus arteriosus typically lead to left-sided chamber enlargement.[2] Unroofed CS, a rare congenital anomaly presenting with partial or complete absence of the roof of the CS, is highly associated with PLSVC.[3,4] In our case, color Doppler and CT ultimately confirmed the correct diagnosis of PAPVR and excluded alternative shunt lesions. Lastly, although the patient did have a pulmonary embolism, its size and location were felt to be insufficient to have caused this degree of right heart enlargement and it was felt to be an incidental finding. The finding of right heart enlargement on a prior TTE years earlier further supports this conclusion.

Scimitar syndrome is a rare congenital heart defect characterized by PAPVR of one or all right-sided pulmonary veins into the IVC. This often results in right lung hypoplasia of varying degrees with

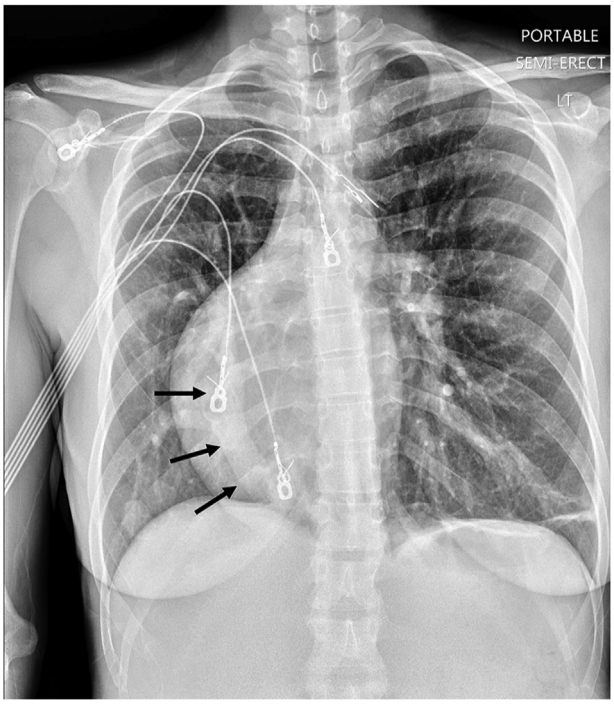

Figure 3 Chest x-ray demonstrates dextrocardia overlying a double-density linear structure representing the PAPVR (*arrows*) draining into the IVC, consistent with scimitar syndrome.

associated cardiac dextroposition.[5] The anomaly has an estimated prevalence of 1 to 3 per 100,000 births.[6] The presence of symptoms and age at presentation typically depend on the number of pulmonary veins that anomalously drain as well as the presence of associated congenital heart disease lesions. The infantile form can present with new heart failure, recurrent pulmonary infections, and severe pulmonary hypertension in children less than 1 year old and is associated with the highest mortality.[7] Conversely, the adult form typically has a milder course, with a lower incidence of right pulmonary hypoplasia and symptomatology but comparable rates of respiratory infection.[8] Transthoracic echocardiography was essential in establishing a diagnosis of scimitar syndrome. Compared with children, visualization of the pulmonary veins on TTE is often limited in adults due to their body size. In some cases of scimitar syndrome, the inferior pulmonary veins can be imaged using the coronal and sagittal subcostal imaging planes. However, in most adults imaging is typically limited, and the diagnosis is suggested by unexplained right heart enlargement. In this setting, it is crucial for the imager to look for congenital shunts. Transesophageal echocardiography, cardiovascular CT, and magnetic resonance images are more sensitive for the detection of PAPVR and should be considered if TTE findings are inconclusive and clinical suspicion remains high.

Surgical repair for scimitar syndrome is indicated in patients with significant left-to-right shunt, which is characterized by right heart enlargement. In severe cases, patients can develop right-sided heart failure, progressive pulmonary hypertension, exercise intolerance, and new-onset atrial and ventricular arrhythmias.[1] Surgical repair of symptomatic scimitar syndrome can be done by baffle repair, creating a tunnel-like connection, or direct pulmonary venous rerouting of the scimitar vein to the left atrium.[5] Patients with recurrent respiratory infections should be evaluated for right lung sequestration, prompting the need for potential right lobectomy or transcatheter coil embolization of aberrant arterial collaterals to these areas. Given the right heart enlargement, this patient likely has a significant left-to-right shunt with QP:QS ratio >1.5, which meets the conventional criteria for surgical intervention.[1] Our patient has a challenging clinical picture given their coexisting cancer diagnosis, and for now surgery has been put on hold. Persistent left-sided vena cava is the most common thoracic venous malformation, with a reported prevalence of up to 0.2% to 3.0% in the general population.[8] Persistent left-sided vena cava occurs due to failure of the left anterior cardinal vein to regress during embryological development.[9] If regression does not occur, a left-sided superior vena cava (SVC) persists, draining into the right atrium most often through the CS. The "low left atrial pressure theory" suggests that in the presence of other intracardiac anomalies, a left-sided SVC fails to obliterate due to insufficient left atrial pressure or left lung development.[8] In left-to-right shunt lesions where right-sided atrial pressure becomes relatively greater than left, like ASD or PAPVR, this theory may explain the coincidence of both venous malformations.

Persistent left-sided vena cava should be suspected when a left subclavian pacemaker electrode abruptly progresses caudally into the CS on chest x-ray rather than across the midline into a right-sided SVC before entering the right atrium. Transthoracic echocardiography typically reveals unexplained dilation of the CS; the pathognomonic feature is passage of microcavitations to the CS prior to the RV on bubble study using a left-arm IV; this should typically be performed in either the parasternal long-axis view or a modified, low apical 4-chamber view in which the CS is visualized.

Failure to recognize PLSVC can result in serious complications during vascular access, particularly CS dissection, thrombosis, or perforation.[9,10] Coronary sinus enlargement, a common outcome of PLSVC, can lead to AV node or His bundle compression in the triangle of Koch and subsequent atrial fibrillation or other supraventricular tachyarrhythmias.[11] This patient received neoadjuvant chemotherapy with 4 cycles of doxorubicin and cyclophosphamide peripherally, avoiding potential complications related to central venous catheter placement. As our patient may require central venous catheter placement for future infusions, understanding of their anatomy will be important for preventing future associated complications related to their PLSVC.

Approximately 70% of scimitar syndrome patients have an associated ASD, most commonly secundum type, with sinus venosus defects being less common.[5,7] Patent ductus arteriosus and ventricular septal defect are also common defects in this population.[5,7,12] The association of scimitar syndrome and PLSVC—a condition in which a distinct left-sided SVC drains directly into the CS—is extremely rare, with only a few reported cases.[13,14]

CONCLUSION

We report a young woman who was diagnosed with right-sided PAPVR with scimitar syndrome and PLSVC, which were both diagnosed incidentally by TTE. Our report highlights the importance of careful interrogation for shunt lesions on TTE whenever unexplained right heart enlargement is present. Additionally, the presence of unexplained CS dilation should raise suspicion for PLSVC, which can be confirmed with saline contrast bubble study with left-arm injection. Knowledge of this variant in thoracic anatomy is essential for preventing complications of a left subclavian vein approach during cardiac device and central line placement.

ETHICS STATEMENT

The authors declare that the work described has been carried out in accordance with The Code of Ethics of the World Medical Association (Declaration of Helsinki) for experiments involving humans.

CONSENT STATEMENT

The authors declare that since this was a non-interventional, retrospective, observational study utilizing de-identified data, informed consent was not required from the patient under an IRB exemption status.

FUNDING STATEMENT

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DISCLOSURE STATEMENT

The authors report no conflicts of interest.

SUPPLEMENTARY DATA

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