An Unusual Cause of Dyspnea Diagnosed Late in Life
Severe Pulmonary Hypertension Resulting From Isolated Anomalous Pulmonary Venous Connection

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A 67-year-old US military veteran was referred to our clinic for evaluation of progressive dyspnea on exertion over the previous 2 years. His medical history was significant for systemic hypertension, obstructive sleep apnea, and the absence of primary lung disease, significant tobacco use, or coronary artery disease. At the time of consultation, his peripheral blood oxygenation saturation was 85%, hepatojugular reflux and lower-extremity edema were noted, and 6-minute walk distance was 34 m.

Transthoracic echocardiography demonstrated normal left ventricular systolic structure and function, a severely dilated right ventricle, and a tricuspid regurgitant jet velocity of 4.85 m/s, indicating an estimated pulmonary artery systolic pressure of 94 mm Hg. However, an elevated flow velocity of 65 cm/s was detected by Doppler interrogation of the posterior aspect of the right atrium adjacent to the interatrial septum. To investigate this further, transesophageal echocardiography was performed, which demonstrated a 1.2-cm communication between the right upper pulmonary vein and the superior vena cava (Figure 1A) and a respirational bidirectional shunt through this lesion (Figure 1B and Video I in the online-only Data Supplement). Three-dimensional echocardiography (Figure 2A and Video II in the online-only Data Supplement) and multislice 3-dimensional reconstructive computed tomographic angiography (Figure 2B) characterized the defect further and confirmed a normal anatomic origin and insertion of the right upper pulmonary vein at the right upper lobe of the lung and left atrium, respectively.

Partial anomalous pulmonary venous connection (PAPVC) describes a rare congenital disease present in up to 0.7% of the general population and is characterized most often by the terminal insertion of a pulmonary vein into the right atrium or superior vena cava in conjunction with a sinus venous atrial septal defect.1 The present case illustrates an isolated anomalous pulmonary vein without an associated atrial septal defect. This anomaly is believed to result from failure of the splanchnic plexus–derived pulmonary vasculature to separate completely from the right common cardinal vein, which is the embryological precursor of the superior vena cava. Thus, the patient’s anatomic anomaly is distinct from the conventional sinus venous atrial septal defect–associated PAPVC because of the unroofing of the common wall between the right upper pulmonary vein and the sinus venosus myocardium, allowing for the superior region of the atrial septum to remain intact.2

Isolated PAPVC has been described previously in the pediatric literature but is an exceedingly uncommon diagnosis in adults.1 We believe this patient’s adulthood presentation is explained, in part, by right-to-left shunt pathophysiology that occurred, ultimately, as a result of severe pulmonary arterial hypertension from PAPVC-dependent increased pulmonary blood flow, chronic systemic hypertension, and obstructive sleep apnea. Surgical closure of the defect with the Warden procedure, which creates a baffle separating the pulmonary from the venous return, is the preferred treatment strategy. However, if surgery is not an option, catheter-based interventions with vascular occlusion coils have been reported.3 Notable potential complications of these procedures include superior vena cava stenosis and pulmonary edema because of instrumentation injury resulting in obstruction of the repaired pulmonary vein.

In this case, severe right ventricular systolic dysfunction (ejection fraction, 28%), severe pulmonary hypertension, and complex PAPVC anatomy conspired to elevate the patient’s estimated procedural risk, thereby favoring a pharmacotherapeutic treatment strategy. Various medical therapies to treat symptomatic, severe pulmonary hypertension in association with congenital heart disease have been reported, including phosphodiesterase type-V inhibitors, endothelin receptor antagonists, calcium channel receptor antagonists, and prostacyclin analogs.4 In the present case, failure to abrogate the

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patient’s severe pulmonary hypertension or symptom burden after therapy with maximum-dose phosphodiesterase type-V inhibition resulted in initiation of parenteral prostacyclin replacement therapy, which improved the patient’s shortness of breath and functional capacity substantially and increased his 6-minute walk distance to 330 m.

Although isolated PAPVC is rare, we believe that greater awareness among the practicing clinical cardiology community is necessary to prevent the delayed or missed diagnosis of these and other potentially important extracardiac circulatory anomalies in patients with otherwise unexplained pulmonary hypertension and heart failure symptoms.

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Figure 1. Echocardiographic capture of an isolated anomalous right upper pulmonary vein (RUPV). A, Transesophageal echocardiographic image acquired at 30° caudal demonstrates unroofing of an enlarged RUPV that results in a 1.2-cm communication (*) between the RUPV and superior vena cava (SVC). B, Doppler color imaging demonstrates respirophasic bidirectional blood flow through the RUPV. LA indicates left atrium.

Figure 2. Three-dimensional characterization of an anomalous right upper pulmonary vein (RUPV)-superior vena cava (SVC) communication. A, Still frame image acquired during 3-dimensional transesophageal echocardiography demonstrates contiguous blood flow between the right upper pulmonary vein (outlined by arrowheads) and SVC. Asterisks (*) designate the margins of the anomalous channel between these structures. B, Multislice 3-dimensional reconstructed computed tomographic angiography (acquired at right anterior oblique of 84°, caudal of 31°) reveals an abnormal RUPV–SVC communication (provided at increased magnification in the inset) with normal insertion of the RUPV into the left atrium (LA). C, Multislice 3-dimensional reconstructed computed tomographic angiography shows a superior course of the anomalous RUPV from the lung parenchyma without meandering before communication with the SVC. Asterisk (*) indicates RUPV. IVC indicates inferior vena cava; and RA, right atrium.
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