Never too old for congenital heart disease: sinus venosus atrial septal defect with anomalous pulmonary venous return in an octogenarian

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Abstract: We report a rare case in an 86-year-old woman with symptoms of exercise intolerance, fatigability, worsening lower extremity edema, and normal ejection fraction on echocardiographic examination who presented with a presumptive diagnosis of heart failure with preserved ejection fraction (HFpEF). Hemodynamic studies revealed that she had normal left-sided filling pressures, mildly elevated pulmonary pressures with normal pulmonary vasculature resistance, and evidence of right ventricular dysfunction. Significant shunting was also detected with a step-up blood oxygen saturation from superior vena cava to right atrium and a calculated pulmonary-to-systemic blood flow ratio of 3.9. Contrast-enhanced multidetector cardiac computed tomography confirmed the presence of a patent foramen ovale, a sinus venosus atrial septal defect, and 3 anomalous pulmonary venous communications to the right atrium and superior vena cava. We hereby present one of the oldest diagnosed cases of sinus venosus defect with anomalous pulmonary venous return as a rare cause of recent-onset dyspnea, volume overload, and functional intolerance in an 86-year-old woman with an initial misdiagnosis of HFpEF.

Keywords: anomalous pulmonary venous return, atrial septal defect, sinus venosus defect, heart failure.

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We report the unusual case of an 86-year-old woman with a medical history of gastroesophageal reflux disease, osteopenia, and chronic back pain who was an avid tennis player before symptom onset and who was evaluated at our center after experiencing 6 months of progressive exercise intolerance, fatigability, worsening lower extremity edema, and abdominal bloating. Her medications included a proton pump inhibitor, calcium and vitamin D supplements, and denosumab (a receptor activator of nuclear factor κ -B ligand inhibitor). Her disease course was also remarkable for refractory atrial fibrillation, which ultimately required atrioventricular nodal ablation and pacemaker implantation. The patient had no previous cardiac imaging.

Two-dimensional echocardiography at the onset of symptoms revealed a small left ventricular cavity with normal wall thickness and an ejection fraction of 60%–65%. The right ventricle (RV) was moderately dilated with moderate hypokinesis; septal flattening was present during diastole consistent with RV volume overload. Although diastolic dysfunction could not be assessed because of atrial fibrillation, significant bi-atrial enlargement was present. On the basis of her clinical and echocardiographic findings, she was given a presumptive diagnosis of heart failure with preserved ejection fraction (HFpEF). Serum protein electrophoresis demonstrated an immunoglobulin A κ monoclonal gammopathy with an abnormal serum light-chain ratio (κ predominant). She was referred for endomyocardial biopsy and right heart catheterization to evaluate for possible infil-

trative cardiomyopathy (i.e., amyloidosis). Her hemodynamic characteristics (Table 1) revealed normal pulmonary capillary wedge pressure (PCWP) of 12 mmHg, mildly elevated pulmonary pressures (mean pulmonary artery pressure, 26 mmHg), and evidence of right ventricular dysfunction (right atrial [RA] pressure of 12 mmHg and hemodynamic Kussmaul's sign). Surprisingly, a significant oxygen saturation step-up was noted between the central veins and the mid-RA, indicating substantial shunting with a calculated pulmonary-to-systemic blood flow ratio (Qp/Qs) of 3.9 (superior vena cava, inferior vena cava, RA, and pulmonary artery blood oxygen saturation of 53.2%, 49.2%, 85.6%, and 85.3%, respectively; Table 1). Indirect Fick systemic (Qs) cardiac output was calculated at 2.75 L/min, with a resultant calculated Qp of 10.7 L/min. Given this Qp, the calculated PVR was 1.3 Wood units. Contrast-enhanced multidetector cardiac computed tomography (MDCT) confirmed the presence of a patent foramen ovale, a sinus venosus atrial septal defect (SV-ASD), and 3 anomalous pulmonary venous communications to the RA and superior vena cava (Fig. 1). Endomyocardial biopsy results were negative for amyloid. After discussion with the cardiology and cardiothoracic surgical teams, the patient opted for conservative management and currently has New York Heart Association class II symptoms while receiving moderate-dose diuretic therapy.

Anomalous pulmonary venous return (APVR) is a congenital pulmonary venous anomaly that is often diagnosed in childhood, with

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Table 1. Right heart catheterization hemodynamic parameters and shunt run

Hemodynamic parameter	Value	Anatomic measuring point	Oxygen saturation, %
Right atrial pressure	12 mmHg	Superior vena cava (high)	53.2
Right ventricular pressure (systolic/end diastolic)	44/10 mmHg	Right atrium (high)	60.2
Pulmonary pressure (systolic/diastolic/mean)	47/15/26 mmHg	Right atrium (mid)	85.6
Pulmonary capillary wedge pressure	12 mmHg	Right atrium (low)	72.8
Pulmonary vascular resistance, systemic cardiac output (indirect Fick) ^a	1.3 Wood units, 2.75 L/min	Right ventricle (mid)	87.1
Systemic cardiac index (indirect Fick) ^a	1.85 L/min/m ²	Pulmonary artery (distal, right)	85.3
Heart rate	80 bpm	Inferior vena cava (high)	49.2
Systemic mean arterial pressure	97 mmHg	Systemic arterial	97.0

^a Indirect Fick formula: $[135 \times (\text{body surface area})] / [1.36 \times 10 \times \text{hemoglobin} \times (\text{arterial oxygen saturation} - \text{mixed venous oxygen saturation})]$, where mixed venous oxygen saturation is estimated by a weighted average of superior vena cava (SVC) and inferior vena cava (IVC) oxygen saturations: $(3 \times \text{SVC oxygen saturation} + 2 \times \text{IVC oxygen saturation}) / 5$.

occasional cases first manifesting in adulthood. It is characterized by abnormal drainage of pulmonary veins into the right-sided circulation, creating a partial left-to-right shunt. APVR is associated with SV-ASD defect in 80%–90% of cases and with ostium primum ASD in 10%–15% of cases.^{1–3} Conversely, 10% of patients with an ASD also have APVR.⁴ Dyspnea, exercise intolerance, palpitations, and lower extremity edema are the usual presenting symptoms in adulthood. Echocardiography, MDCT, and cardiac magnetic resonance imaging are noninvasive diagnostic modalities used to assess the anatomy and abnormal communications. MDCT's rapid scan acquisition, with high spatial resolution and wide anatomic coverage,

allows accurate anatomical assessment of both APVR and SV-ASD.^{5,6} Catheter-based angiography provides information on the hemodynamic characteristics and oxygenation within the cardiac chambers and allows volumetric quantification of the ASD. Patients with asymptomatic APVR undergo periodic follow-up, and medical therapy is not routinely indicated.

Earlier studies have reported that patients presenting with symptoms at an early age tend to have more severe disease and multiple congenital malformations requiring surgical intervention. Conversely, APVR diagnosed during late adulthood represents subclinical or mild disease with few associated abnormalities, usually presenting as left

Figure 1. A, Sagittal four-chamber view demonstrating superior sinus (black arrows) venosus defect connecting the left and the right atrium. B, Axial image representing anomalous pulmonary vein (white arrows) draining into the superior vena cava (SV). AA: ascending aorta; LA: left atrium; LV: left ventricle; PA: pulmonary artery; RA: right atrium; RV: right ventricle.

upper lobe APVR or isolated right upper lobe APVR without sinus venosus atrial septal defect.⁷ Furthermore, contrast-enhanced MDCT technology is underutilized during diagnostic evaluation of such cases. In this regard, our case represents an atypical presentation of APVR with significant sinus venosus defect that remained quiescent until old age and was comprehensively evaluated using MDCT imaging.

Over time, high flow through the pulmonary circuit from left-to-right shunting can cause pulmonary hypertension and subsequent right ventricular compromise. The quantitative flow through the pulmonary circuit is directly correlated with the risk of subsequent RV dysfunction and heart failure, which is the major determinant of symptom onset and progression. In this setting, volume overload is managed with diuretics. Pulmonary vasodilators can be considered, but testaments to their efficacy are relegated to case reports only.^{8,9} Arrhythmias should be treated accordingly. Definitive treatment for APVR is surgical repair. In the pediatric population, a Qp:Qs of >1.5 is often cited as an indication for surgery. Indications for surgical interventions in adults are not as clearly defined.¹⁰ However, adult patients who are appropriate surgical candidates with an elevated Qp:Qs, signs of RV dilation or compromise, and burgeoning pulmonary hypertension should all be considered for surgical repair. In our patient's case, even after accounting for the moderate expected increase in shunt fraction with age, her normal PVR is quite remarkable after 86 years of significant left-to-right shunting.

We report a sinus venosus defect with anomalous pulmonary venous return as a rare cause of recent-onset dyspnea, volume overload, and functional intolerance in an 86-year-old woman who received an initial misdiagnosis of HFpEF. This is one of the oldest reported cases of sinus venosus defect in the literature.

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