Diagnostic and therapeutic challenges in identifying a rare pulmonary arterial hypertension variant associated with portal hypertension and multiple abdominal vascular malformations

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Pulmonary arterial hypertension (PAH) in general is a scarce extraordinarily complex disease entity that remains still incurable, resulting in right ventricle (RV) failure in the majority of patients, potentially leading to premature death [1, 2]. The coexistence of PAH with portal hypertension (PH) is commonly called portopulmonary hypertension (PoPH), and the percentage of patients newly diagnosed with PoPH is thought to be as high as 15% of all patients with PAH and is steadily increasing [3]. However, recent studies indicate that treating PoPH with PAH-specific therapies may be beneficial, as reflected in improved functional and hemodynamic parameters and reduced disease severity. It also enables liver transplantation (LT) [4].

We present the case of a 28-year-old female Ukrainian refugee with a clinically established diagnosis of PH associated with PAH in World Health Organization functional class II. The patient had previously participated in the PAH therapeutic program in Kyiv, receiving sildenafil monotherapy. Upon admission, her general condition was good, with a 6-minute walk test of 610 m; a resting electrocardiogram showed sinus rhythm, a dextrogram, and features of RV hypertrophy. During hospitalization, the diagnosis of PoPH was expanded. Right heart catheterization was performed, confirming severe PAH. Mean pulmonary artery pressure was 70 mm Hg; pulmonary capillary wedge pressure was 11 mm Hg; right atrial pressure was 9 mm Hg; the cardiac index was 3.7; and pulmonary vascular resistance was 8.1 WU.

Based on ventilation-perfusion scintigraphy, the thromboembolic etiology of PAH was excluded. Transthoracic echocardiography showed RV enlargement (Figure 1A), D-sign, tricuspid annular plane systolic excursion ≥20 mm (Figure 1B), mild tricuspid regurgitation with significantly elevated tricuspid regurgitation peak gradient ~100 mm Hg (Figure 1C) and left ventricular ejection fraction ~66%. Based on magnetic resonance imaging (Figure 1D–F), the following changes were diagnosed: agenesis of the portal vein trunk and its hepatic branches, joint confluence of the superior mesenteric vein and the dilated splenic vein through the collaterals to the left renal vein, and departure of the left common iliac vein into the left renal vein (so-called Abernethy malformation), as well as the presence of aneurysms of the splenic and hepatic arteries. In addition, a past splenic infarction, and a horseshoe kidney supplied by two renal arteries were visualized. The focal liver lesion seen earlier was located subcapsularly in segment VII and currently had dimensions of approximately 78 × 64 × 65 mm (LR × AP × CC). The histopathological examination with a fine needle aspiration biopsy showed no neoplastic tissue.

At the time of writing this clinical vignette, the patient remained in class A (Child–Pugh scale) of liver cirrhosis. During hospitalization in the Department of Cardiology, therapy with

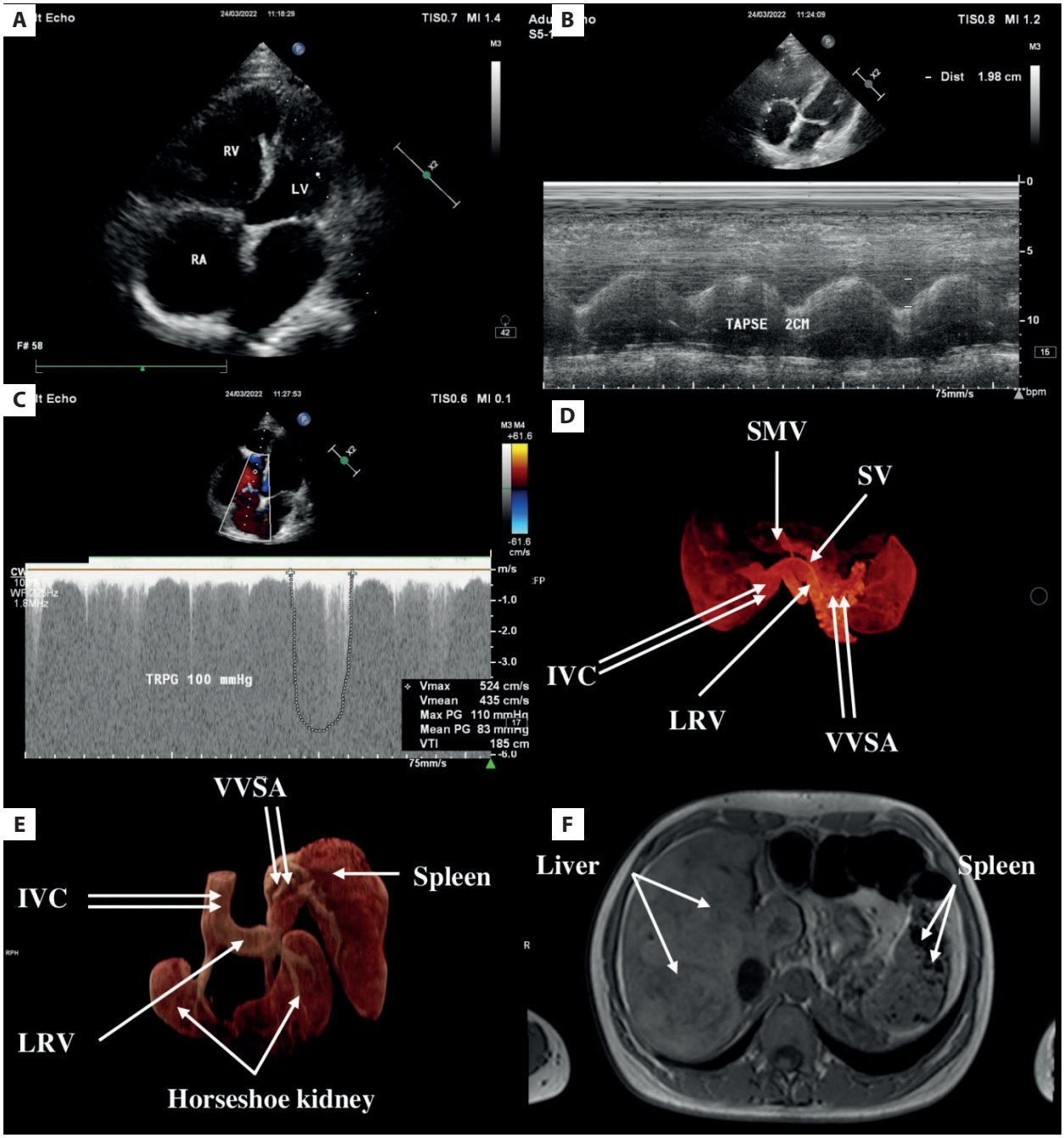

Figure 1. A. Echocardiography — TTE 2D modified 4C: visible enlargement of the RA and RV cavities with displacement and bulging of the RV towards the LV. **B.** Echocardiography — TTE M-mode: TAPSE calculation. **C.** Echocardiography — TTE CD: significantly elevated TRPG ~100 mm Hg. **D.** Magnetic resonance 3D volume rendering of T1-weighted contrast-enhanced subtraction images: SMV (single solid arrow) connects with the SV (single dropped arrow) and flows through the VVSA (double solid arrow) into the IVC (double dashed arrow) through the dilated LRV (single dashed arrow); neither the true trunk of the portal vein in the area of the hepatic hilum nor its intrahepatic branches are visible. **E.** Magnetic resonance 3D volume rendering of T1-weighted contrast-enhanced subtraction images: numerous varicose-shaped venous vessels of the collateral circulation are visible in the area of the splenic hilum (double solid arrow), draining into the significantly dilated LRV (single dashed arrow) and then to the IVC (double dashed arrow); horseshoe kidney also visible (double dropped arrow). **F.** Magnetic resonance — MRI axial T1 weighted image: heterogeneous, nodularly remodeled liver parenchyma (dashed arrows) and spleen with dilated venous lacunae (solid arrows)

Abbreviations: 4C, four-chamber; CD, color doppler; IVC, inferior vena cava; LRV, left renal vein; LV, left ventricle; RA, right atrium; RV, right ventricle; SMV, superior mesenteric vein; SV, splenic vein; TAPSE, tricuspid annulus peak systolic excursion; TRPG, tricuspid regurgitation peak gradient; TTE 2D, two-dimensional transthoracic echocardiography; VVSA, varicose-shaped veins around the spleen

high doses of sildenafil was continued, and typical pharmacological treatment for RV failure was introduced. An attempt to include bosentan proved unsuccessful, and the patient experienced severe headaches (similar to those after ambrisentan). Earlier, we had also made an unsuccessful attempt to include iloprost (allergic symptoms). The patient was discharged for ongoing care in good general condition.

In this group of patients, early PoPH diagnosis seems to be of pivotal importance; it has a huge impact on the prognosis and potential LT candidate selection. PAH-targeted therapy further represents the treatment of choice [5]. Numerous concomitant vascular system defects, liver cirrhosis, and PAH remain a diagnostic and therapeutic challenge.

Article information

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