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INTERMEDIATE

IMAGING VIGNETTE: CLINICAL VIGNETTE

Congenitally "Inverted" Pulmonary Valve in Tetralogy of Fallot

When Nature Falters

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ABSTRACT

A cyanotic neonate with tetralogy of Fallot was found to have a congenitally inverted pulmonary valve. Diagnosis was made via echocardiography and cardiac catheterization. The valve opened retrograde into the right ventricle, which allowed severe regurgitation and prevented anterograde flow. This report is the first description of this anomaly in medical literature. (**Level of Difficulty: Intermediate.**) (J Am Coll Cardiol Case Rep 2020;2:544–6) Published by Elsevier on behalf of the American College of Cardiology Foundation. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

A full-term neonate born to a 30-year-old woman was noted to be dusky at birth. Oxygen saturation was between 70% and 75% without improvement on supplemental oxygen. Prostaglandins were initiated, and the neonate was transferred to Mount Sinai Medical Center for a cardiac evaluation.

Transthoracic echocardiography revealed aortic over-riding, a large anterior malalignment ventricular septal defect (VSD), absent right pulmonary artery, left pulmonary vein stenosis, and aortopulmonary collaterals. The main pulmonary artery (MPA) connected to a hypoplastic left pulmonary artery with no anterograde flow across the pulmonary valve. There was severe pulmonary regurgitation (**Figure 1**). Cardiac catheterization was performed for delineation of pulmonary arteries and collaterals. Two large aortopulmonary collaterals supplying the right and left lungs were seen. Selective injection showed a third collateral to the hypoplastic left lung, which supplied the hypoplastic MPA retrograde, with contrast regurgitating into the right ventricle (RV). However, RV injection revealed no anterograde filling of the MPA (Video 1). Because of this unusual physiology, a repeat echocardiogram was performed that revealed that the pulmonary valve was opening in the reverse direction (Video 2). This abnormal valve had chordal attachments to a papillary muscle in the outflow tract (**Figure 1**).

Because of the predominant supply of pulmonary blood flow through collateral vessels, the patient remained in a well-balanced state with oxygen saturations between 80% and 85%. At 5 months, he underwent surgical unifocalization of the right-sided collaterals and creation of a RV-to-pulmonary artery conduit with closure of the VSD. His post-operative course was complicated by pulmonary hypertension that required serial cardiac catheterizations and medications. He also had left lung pneumonectomy and tracheostomy for

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The authors attest they are in compliance with human studies committees and animal welfare regulations of the authors' institutions and Food and Drug Administration guidelines, including patient consent where appropriate. For more information, visit the *JACC: Case Reports* author instructions page.

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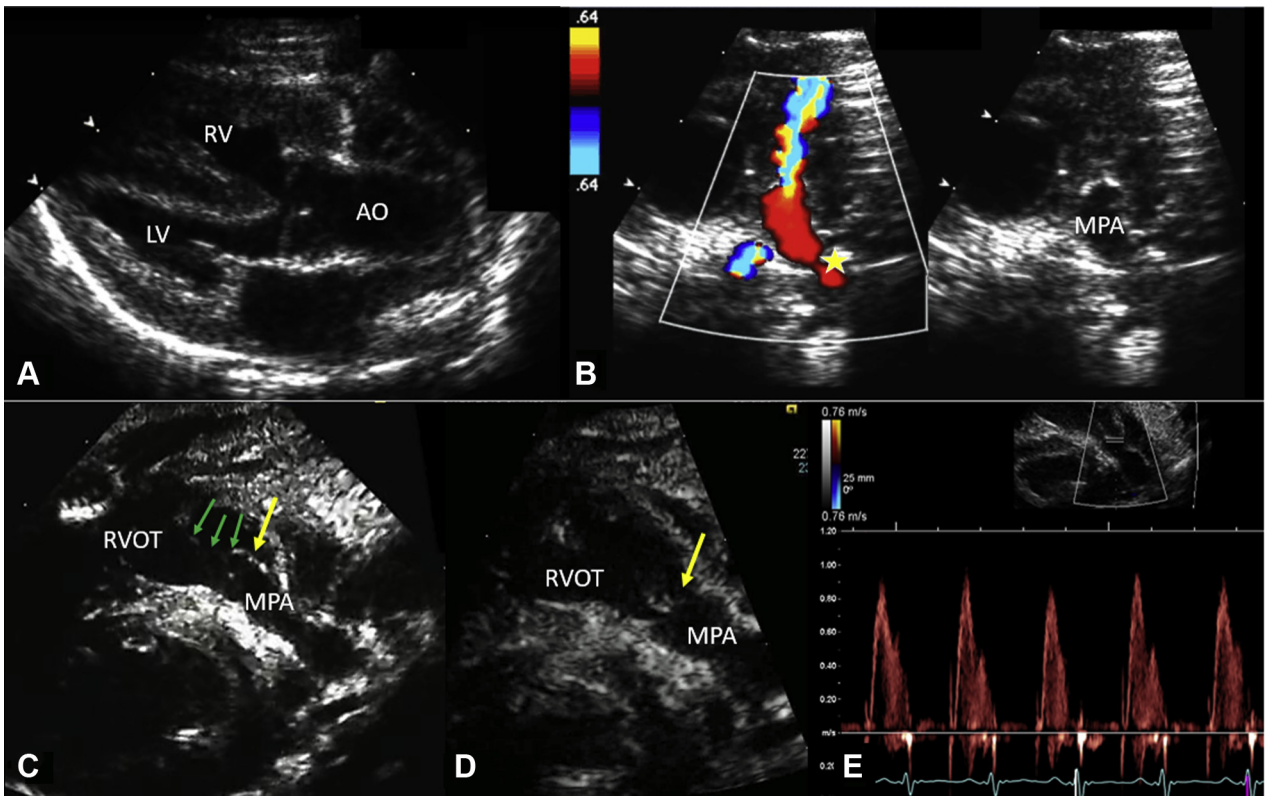
FIGURE 1 **Initial Transthoracic Echocardiography Images of the Inverted Pulmonary Valve in Tetralogy of Fallot**



(A) Parasternal long-axis image showing an over-riding aorta and ventricular septal defect. There is aortic-mitral fibrous continuity. **(B)** Parasternal short-axis color comparison shows retrograde flow toward the right ventricle that is consistent with regurgitation. The main pulmonary artery (MPA) continues into the left pulmonary artery **(yellow star)**. The right pulmonary artery is not seen connecting with the MPA. **(C)** Two-dimensional image from a parasternal long-axis view showing the pulmonary valve leaflets **(yellow arrow)** opening toward the right ventricular outflow tract (RVOT) in diastole. Chordal attachments of the valve to the papillary muscle in the RVOT can be seen **(green arrows)**. **(D)** Two-dimensional image from a parasternal long-axis view showing closure of the valve leaflets **(yellow arrow)** in systole that prevents anterograde flow into the main MPA. **(E)** Pulse wave Doppler at the pulmonary valve with the electrocardiogram strip showing flow above the baseline (toward the right ventricle [RV]) during diastole and no flow in systole. AO = aorta; LV= left ventricle.

persistent hemoptysis in the setting of ineffective left-sided pulmonary blood flow. Genetic testing revealed duplication in chromosome 17q11, a variant of unknown significance. At his last follow-up, at age 6 years, he remains mechanically ventilated via tracheostomy, with well-controlled pulmonary hypertension.

Tetralogy of Fallot (characterized by infundibular hypoplasia and stenosis, malalignment VSD, RV hypertrophy, and an over-riding aorta) has a varied anatomic spectrum, including pulmonary stenosis, atresia, and absent pulmonary valve syndrome. Clinical presentation and management depend largely on the anatomy of the pulmonary valve and arterial architecture (1–3).

The pathophysiology in this patient was similar to that of tetralogy of Fallot with pulmonary atresia with absent anterograde flow from the RV to the pulmonary arteries. Despite the presence of patent pulmonary valve leaflets, their reverse orientation resulted in "competence" of the closed valve in ventricular systole (simulating atresia physiology) and wide valve opening in diastole, thus allowing retrograde RV diastolic filling (pulmonary regurgitation). All pulmonary blood flow was supplied via aortopulmonary collateral vessels.

We report a case of an inverted pulmonary valve in the setting of complex tetralogy of Fallot associated with severe pulmonary artery hypoplasia and multiple aortopulmonary collaterals. To date, there has been no description of a congenitally inverted pulmonary valve in medical literature. This report aims to raise awareness about this rare structural abnormality and its unusual pathophysiology.

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KEY WORDS congenital heart defect, cyanotic heart disease, pulmonary atresia, pulmonary valve, tetralogy of Fallot

APPENDIX For supplemental videos, please see the online version of this paper.