Case Reports

Unrepaired Tetralogy of Fallot with Absent Pulmonary Valve

in a Mildly Symptomatic 16-Year-Old Boy

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Absent pulmonary valve is a rare and severe variant seen in only 3% to 6% of patients with tetralogy of Fallot. Fetuses with this combined condition who survive through birth typically need intervention in infancy or early childhood because of respiratory distress, heart failure, or failure to thrive. We describe the unusual case of a mildly symptomatic 16-year-old boy with these conditions who underwent successful primary repair. Our search of the medical literature yielded fewer than 5 cases of tetralogy of Fallot with absent pulmonary valve (or variants with an absent left pulmonary artery) and survival without repair into later adolescence or adulthood. ***(Tex Heart Inst J 2016;43(6):517-9)***

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Tetralogy of Fallot (TOF) with absent pulmonary valve (PV) is a severe congenital heart disease found in only 3% to 6% of TOF patients.[1] Survival into young adulthood, especially without repair, is quite rare. The median age at time of repair ranges from 9 months to 1.4 years.[2,3] Even with repair during infancy, postoperative mortality rates range from 11% to 30%, and the 20-year survival rate is only 81%.[2] We present the case of a patient with only mild symptoms who underwent surgical repair for this combined condition as an adolescent.

Case Report

In January 2015, a 16-year-old boy with no surgical history presented after the detection of a murmur. He and his parents were from Mexico, where his parents had been told that their son had a grave cardiac condition. Because of mortality rates said to exceed 50% in Mexico, they had been urged not to pursue surgical correction.

The parents had noted no issues with their son's growth, development, or respiratory capabilities throughout his childhood and most of his adolescent years; he merely tired more rapidly than his peers and had occasional fluttering in his chest. He was taking no medications.

On examination, the patient was acyanotic and in no distress. His pulse rate was 75 beats/min; blood pressure, 107/68 mmHg; and oxygen saturation, 100% on room air. For his age, his weight was in the 33rd percentile and his height in the 53rd. Cardiac examination revealed strong, symmetric pulses bilaterally; a grade 4/6 holosystolic murmur at the left upper sternal border with radiation throughout the thorax; and a 3/6 diastolic murmur at the left sternal border. The patient had no hepatomegaly or peripheral edema. A chest radiograph showed moderate cardiomegaly and dilation of the pulmonary artery (PA) (Fig. 1).

A 2-dimensional transthoracic echocardiogram revealed severe pulmonary regurgitation and stenosis (Fig. 2). The main PA was dilated to 5.13 cm (Z score, 9.37) and the left PA to 3.72 cm (Z score, 10.36). The PV leaflets were extremely rudimentary, consistent with the physiology of TOF with absent PV (Fig. 3). The right ventricle was severely dilated with mildly reduced systolic function; left ventricular size and function were preserved. There was no pulmonary hypertension. The tricuspid regurgitation jet was 14 mmHg with a normal septal contour. A large, nonrestrictive ventricular septal defect had predominantly left-to-right flow. The PA enlargement was confirmed on cardiac magnetic resonance images (Figs. 4 and 5), which also revealed normal coronary anatomy and an extremely elevated right ventricular end-diastolic volume of 207.2 mL.

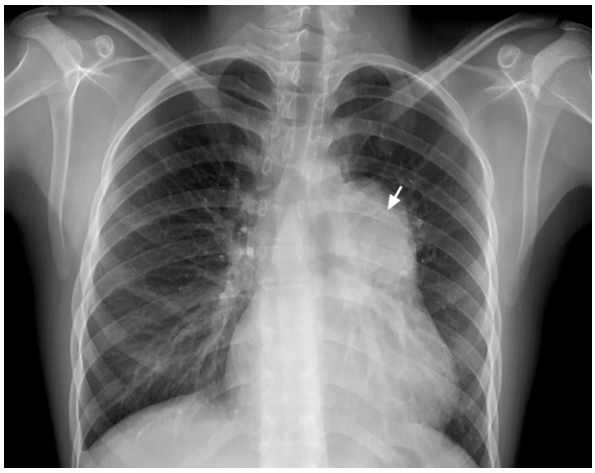

Fig. 1 *Chest radiograph shows an enlarged central pulmonary artery (arrow) and moderate cardiomegaly.*

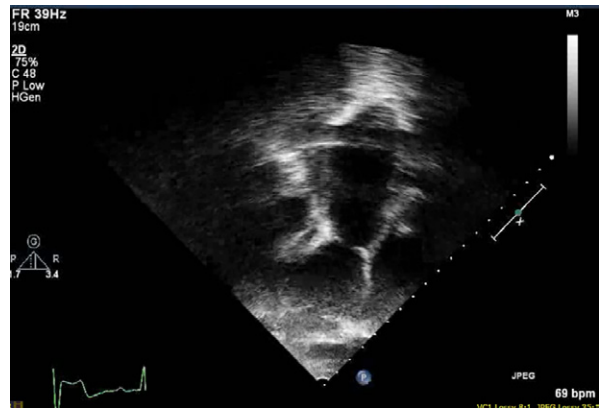

Fig. 3 *Two-dimensional transthoracic echocardiogram shows rudimentary pulmonary valve leaflets, a pulmonary annular diameter of 1.7 cm (Z score, –2.2), and a main pulmonary artery diameter of 5.13 cm (Z score, 9.37).*

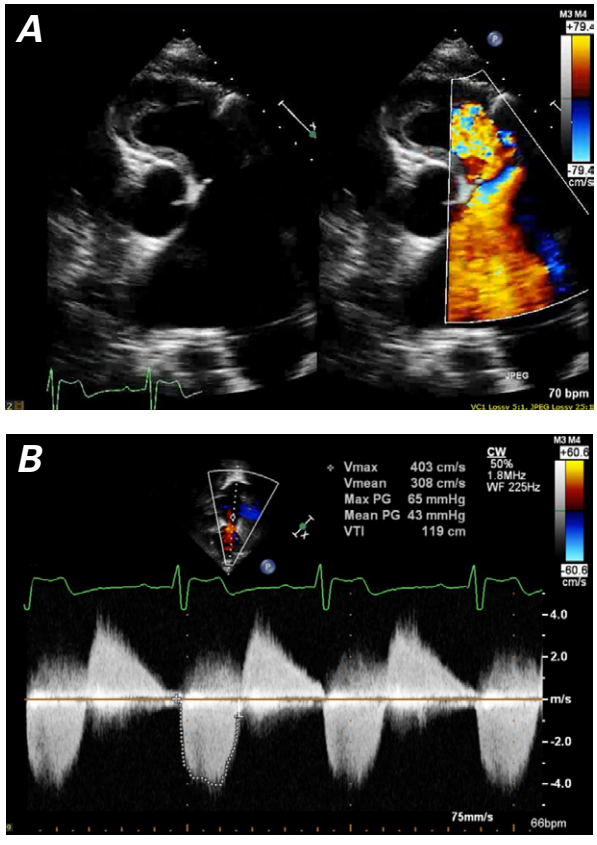

Fig. 2 ***A****) Two-dimensional transthoracic echocardiogram shows rudimentary-to-nearly absent pulmonary valve leaflets; color-flow Doppler mode reveals severe pulmonary regurgitation and stenosis starting below the pulmonary annulus.* ***B****) Continuous-wave Doppler tracing through the right ventricular outflow tract shows pulmonary regurgitation and stenosis (peak gradient, 65 mmHg; mean, 43 mmHg).*

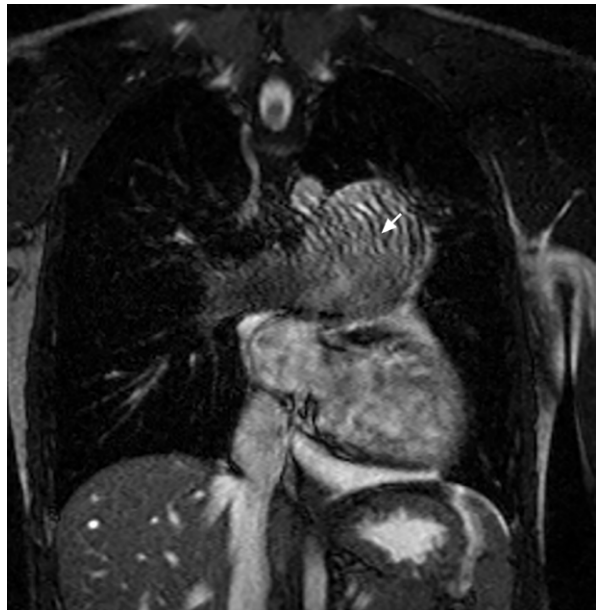

Fig. 4 *Cardiovascular magnetic resonance image shows aneurysmal enlargement of the main pulmonary artery (5.3 × 4.6 cm).*

The patient underwent successful repair with use of a transannular patch and the implantation of a 27-mm Mosaic® bioprosthetic valve (Medtronic, Inc.; Minneapolis, Minn) in the pulmonary position. The native PV had thickened leaflets that had failed to delaminate, and they adhered to the right ventricular wall. The distal PA was profoundly dilated. Despite the exaggerated size of the PAs, we decided not to plicate them when we saw them—the patient had no pulmonary hypertension and was asymptomatic, so the risk of PA rupture or worsening of the aneurysmal dilation was low.

The patient was successfully extubated that same day, needed no supplemental oxygen or reintubation throughout his hospital course, was discharged on hospital day 4, and had a rapid return to normal daily life. As of October 2016, he was doing well in high school and keeping up with his peers during physical activity.

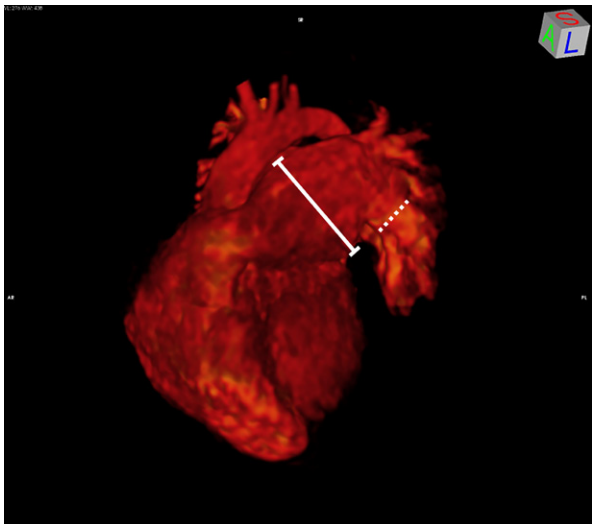

Fig. 5 *Cardiovascular magnetic resonance image (3-dimensional reconstruction) shows enlarged main (solid line) and left (dashed line) pulmonary arteries.*

Discussion

Unrepaired TOF with absent PV is rarely survived into young adulthood. Rudimentary or absent PV is usually also associated with a stenotic pulmonary annulus and aneurysmal dilation of the PAs.[3,4] Prenatal diagnosis carries a poor prognosis.[5,6] In fetuses that survive, morbidity and mortality rates are high, usually because of bronchial compression from highly pulsatile, massive PAs, which leads to bronchomalacia or air-trapping.[7] Typically, patients present early in life with cyanosis, recurrent respiratory distress, failure to thrive, or heart failure.[8] Reports of adults with TOF and absent PV are rare. In one report,[9] a woman in her 30s was diagnosed; unlike our patient, she presented with respiratory symptoms.

Our patient had only minimal symptoms before presentation, making his case unusual. His growth had been unhindered by the congenital anomalies, and he did not have the recurrent respiratory issues typically associated with bronchial tree compression. His anatomy was advantageous in that obstruction at the level of the PV impeded pulmonary overcirculation.

Had our patient presented later in life, pulmonary compromise might have occurred. However, he was fortunate to have well-compensated physiology and no signs of bronchial compression, enabling an uncomplicated intracardiac repair and a rapid return to daily life.

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