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CASE REPORT

Echocardiographic Features of Adult Tetralogy of Fallot with Natural Palliative Correction by Patent Ductus Arteriosus

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A thirty-year-old man with the diagnosis of the tetralogy of Fallot and patent ductus arteriosus was admitted to our hospital because of a syncope. He reported no previous symptoms. We diagnosed adult tetralogy of Fallot, which included all four characteristic anomalies: ventricular septal defect, overriding aorta, pulmonary artery stenosis, and right ventricular hypertrophy. The associated persistent ductus arteriosus and the presence of compensatory arteriovenous communications produced a continuous flow load on the left ventricle, which resulted in moderate left ventricular hypertrophy, but without symptoms of pulmonary congestion or cardiac decompensation. Anatomic diagnosis and hemodynamic assessment were established by transthoracic and transesophageal echocardiography, with incidental finding of a quadricuspid aortic valve. To the best of our knowledge, our case of the adult form of Fallot's tetralogy associated with both patent ductus arteriosus and quadricuspid aortic valve is the first one ever described. It is well known that patients with tetralogy of Fallot who do not undergo operation in childhood have short survival, which depends predominantly on the degree of pulmonary artery stenosis and early development of collateral circulation to the lungs. Long-term persistence of natural aortopulmonary anastomosis with systemic collateral circulation to the lungs and remodeling of the heart, with better hemodynamic balance as well as the presence of mild pulmonary artery stenosis probably enhanced the survival of our patient.

Key words: *aortic valve; echocardiography; heart defects, congenital; patent ductus arteriosus; tetralogy of Fallot*

The tetralogy of Fallot is a congenital heart disease with a developmental defect due to antero-cephalad deviation of the outlet septum, which results in four anatomic features: 1) right ventricular outflow tract obstruction, which may be infundibular, valvular, or a combination of both; 2) nonrestrictive ventricular septal defect; 3) overriding aorta; and 4) right ventricular hypertrophy. Accompanying features can include additional ventricular septal defects, anomalous coronary arteries, right-sided aortic arch, aortic root dilatation, aortic regurgitation, persistent left superior vena cava, and patent ductus arteriosus. When the tetralogy of Fallot is associated with atrial septal defect, the anomaly is referred to as the pentalogy of Fallot (1). In most cases, tetralogy of Fallot is cyanotic heart disease, but it can be acyanotic as well. Since the ventricular septal defect is usually large, with the area as great as that of the aortic valve, both ventricles and aorta have about the same systolic pressures. The most important hemodynamic factor is the ratio between the resistance to flow into the aorta and resistance to flow across the stenotic right ventricular infundibulum. In the acyanotic form of the tetralogy of Fallot, pulmonary stenosis is mild, the resistance to the right ventricular flow is not large, the pulmonary flow may be twice the systemic flow, and arterial oxygen saturation may be normal. Conversely, in the cyanotic form, pulmonary stenosis is very severe and the resistance to the pulmonary flow is significantly increased, causing right-to-left shunting, with arterial desaturation and subsequent polycythemia (2). Although the average life span of a patient with tetralogy of Fallot who does not undergo operation is 12 years, it has been estimated that nearly 11% survive to the age of 20,6% to the age of 30, and only 3% to the age of 40 (3-5). Survival to the seventh decade has been described, but outcome and life span of an unoperated patient is poor because the natural course of the disease is determined by the severity of the obstruction of the right ventricular outflow tract and pulmonary vasculature (6,7). Prolonged survival is presumably related to the anatomic defects, which result in comparatively mild hemodynamic alterations or remarkable physiologic compensatory responses in the patient's pulmonary circulation, or both. We describe a case of acyanotic adult tetralogy of Fallot with patent ductus arteriosus and quadricuspid aortic valve.

Case Report

A 30-year-old man was admitted to our hospital for syncope after physical effort, which was greater than usual. Tetralogy of Fallot and patent ductus arteriosus were diagnosed by cardiac catheterization, right and left ventricular angiography, and aortography at the age of 6. He had reached the third decade of life without previous palliative or corrective operation and without any symptoms. On the physical examination, he was of normal body growth, acyanotic, with blood pressure of 110/80 mm Hg and discrete finger clubbing. On auscultation, continuous murmur with peak intensity at S2, thereafter gradually fading to termination before S1, was best heard in the second left intercostal space and was related to the pulmonary artery stenosis and patent ductus arteriosus. Hematologic findings were normal, including hemoglobin, hematocrit, and platelet count. The total heart size was normal on chest radiography, with concave pulmonary artery segment and elevated apex, giving a "boot shaped" contour. The measurement of arterial partial oxygen pressure indicated O_2 saturation near 88%. Cyanosis was not detectable. Electrocardiogram showed sinus rhythm with a mean QRS-axis +80° and biventricular hypertrophy (Fig. 1A). Continuous ambulatory electrocardiographic examination was not diagnostically helpful. With M-mode and two-dimensional measurements performed from the left parasternal and apical views of the long axis of the heart, we obtained the following echocardiographic variables: aortic root dimension (Ao) 3.8 cm, left atrial dimension (LA) 5.0 cm, right atrial dimension (RA) 5.1 cm, right ventricular end-diastolic dimension (RVDD) 2.2 cm, left ventricular end-diastolic dimension (LVEDD) 5.6 cm, left ventricular end-systolic dimension (LVESD) 3.8 cm, interventricular septal thickness (IVST) 1.4 cm, right ventricular free wall (RVFW) 1.4-1.8 cm, left ventricular posterior wall (LVPW) 1.5 cm, diameter of ventricular septal defect (VSDd) 1.9 cm (Fig. 1B), and LV ejection fraction 61%. Comprehensive Doppler echocardiography assessment revealed mild mitral and significant tricuspid regurgitation as well as normal aortic valve flow. Infundibular and unicuspid valvular pulmonary artery stenosis with a right ventricular outflow orifice diameter of 0.86 cm, determined on the subvalvular level, was revealed on transesophageal basal transverse view of the main pulmonary artery (8). Post-stenotic dilatation was also demonstrated in the proximal pulmonary artery (Fig. 2A). The velocity time integral of 18.6 cm, right ventricular acceleration time of 95 ms, and increased pulmonary valve peak velocity of 1.2 m/s were assessed by pulsed wave Doppler signal, with the sample volume placed at a

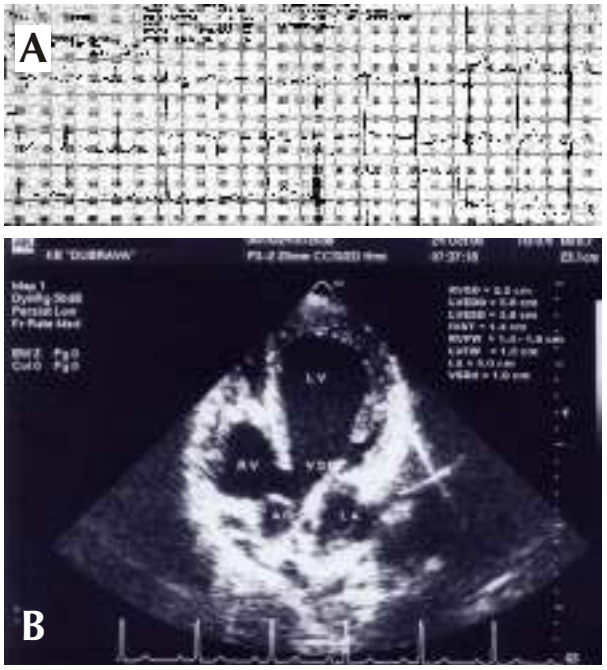

Figure 1. Biventricular hypertrophy in the patient with tetralogy of Fallot and patent ductus arteriosus. **A**. Biventricular hypertrophy recorded by 12-lead electrocardiogram. **B**. Biventricular hypertrophy recorded by two-dimensional echocardiographic tissue harmonic imaging from an apical 5-chamber view.

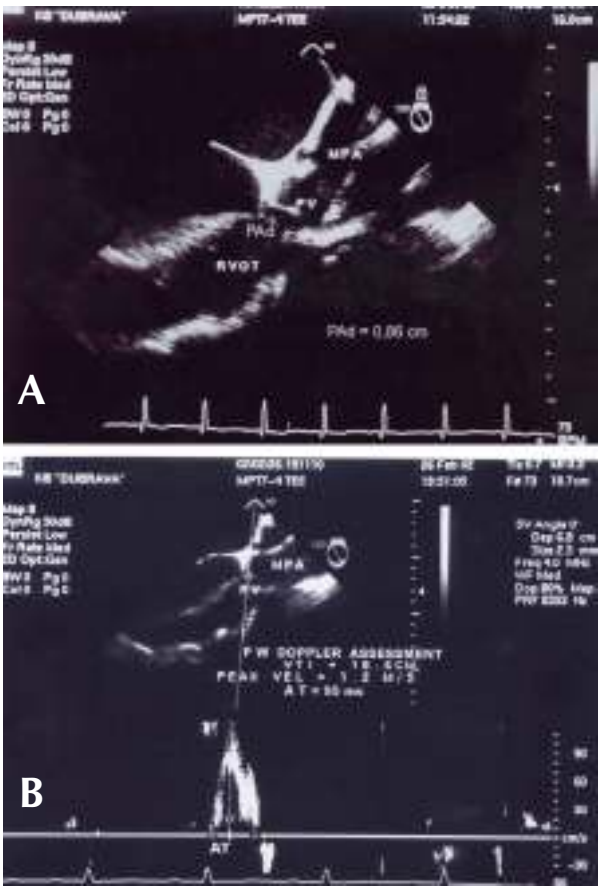

Figure 2. Pulmonary artery stenosis image and hemodynamic measurements in the patient with tetralogy of Fallot and patent ductus arteriosus. **A**. Transesophageal image of the right ventricular outflow tract with infundibular and unicuspid valvular pulmonary artery stenosis and post-stenotic dilatation. The diameter of the pulmonary artery orifice, determined on subvalvular level, was 0.86 cm. **B**. Pulsed-wave Doppler assessment of the pulmonary artery obtained by multiplane transesophageal echocardiography in basal transverse plane position. MPA – main pulmonary artery; PV – pulmonary valve; PAd – pulmonary artery diameter; RVOT – right ventricular outflow tract; VTI – velocity time integral; AT – acceleration time.

precise point just proximal to the pulmonary valve (9,10) and in the same plane (Fig. 2B). The stroke volume and flow per minute of the main pulmonary artery were obtained by the following measurements and formulas: pulmonary artery diameter (PAd) = 0.86 cm; velocity time integral (VTI, distance a column of blood travels with each stroke) = 18.6 cm; cross-sectional area of pulmonary artery annulus (CSA) = $0.785 \times (PAd)^2 = 0.58$ cm^2; stroke volume (SV) = CSA × VTI = 0.58 × 18.6 = 10.8 mL; heart rate (HR) = 74 beats per minute; pulmonary artery flow/min = PA-SV × HR/1000 = 10.8 × 74/1000 = 0.8 L/min; right ventricular acceleration time (RV-AT) = 95 ms; mean pulmonary artery pressure (MPAP) = 90 - (0.62 × RV-AT) = 31 mm Hg.

These parameters indicated reduced relative pulmonary artery stroke volume and flow per minute (Doppler-derived calculations across a stenotic valve produce a "false" increase in the calculated integral because of increases in velocity in the vicinity of an abnormal valve; ref. 11), which demonstrated mild pulmonary artery stenosis with mild pulmonary hypertension (10,11). Right ventricular systolic pressure, estimated by summing the peak pressure of tricuspid regurgitation ($P = 4V^2$) and right atrial pressure was 112 mm Hg. Nonrestrictive and mostly perimembranous large ventricular septal defect and overriding aorta wider than normal were optimally assessed by use of multiplane transesophageal echocardiography, rotating the scanning angle to 120° (Fig. 3A). Using transthoracic and Doppler echocardiography in parasternal long-axis view, we were able to assess the flow through a large ventricular septal defect producing a fairly low velocity jet during the systole (Fig. 3B). This indicated near equalization of the right and left ventricular pressures, with biventricular hypertrophy. Occasionally, patients with adult tetralogy of Fallot have been treated by aortopulmonary shunt as a palliative measure (2), which was in this case naturally solved with a patent ductus arteriosus, well visualized on the two-dimensional echocardio-

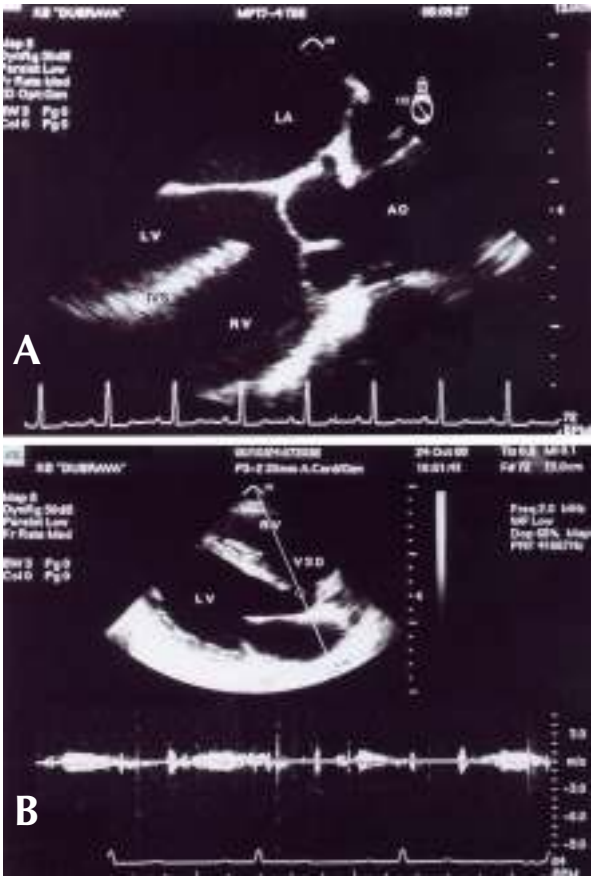

Figure 3. Patient with tetralogy of Fallot and patent ductus arteriosus **A**. Transesophageal echocardiogram of perimembraneous ventricular septal defect, with overriding aorta. **B**. Continuous-wave Doppler assessment of flow through the large ventricular septal defect revealed a fairly low velocity jet during the systole, indicating near equalization of the right and left ventricular pressures. AO – aorta; IVS – interventricular septum; LA – left atrium; LV – left ventricle; RV – right ventricle; VSD – ventricular septal defect.

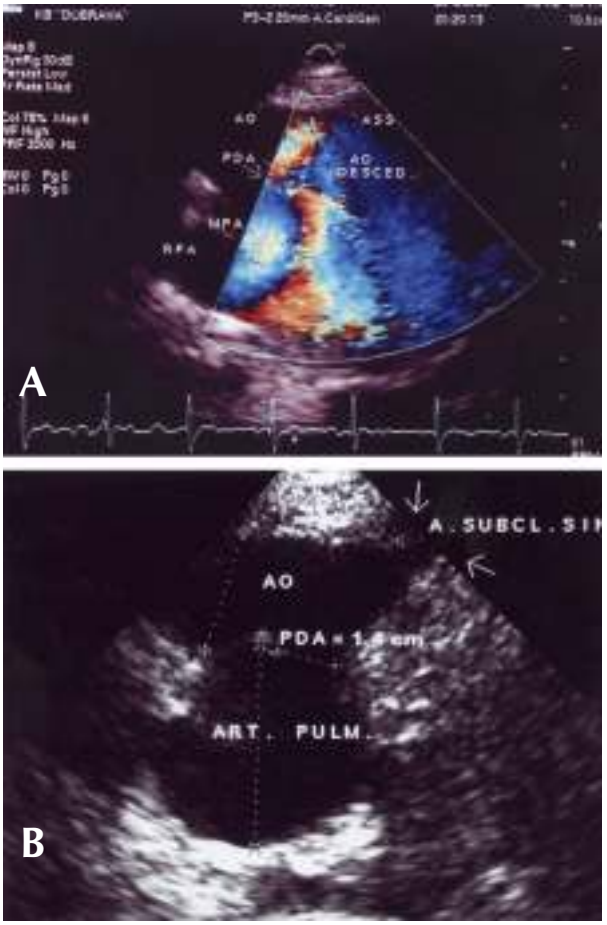

Figure 4. Patient with tetralogy of Fallot and patent ductus arteriosus. **A**. Suprasternal two-dimensional echocardiogram longitudinal to the aortic axis showing the aorta and dilated main pulmonary artery. Color flow Doppler image during systole revealed anterograde flow (colored blue) in the main pulmonary artery and a mosaic jet along the aortopulmonary connection. This turbulent jet was the result of a patent ductus arteriosus. **B**. Patent ductus arteriosus, with maximum distal ductal orifice diameter of 1.4 cm. AO – aorta; ASS – arteria subclavia sinistra (left subclavian artery); MPA – main pulmonary artery; RPA – right pulmonary artery; ART. PULM. – main pulmonary artery; PDA – patent ductus arteriosus.

graphic imaging in the suprasternal long-axis view (Fig. 4A). Color flow imaging during systole revealed anterograde flow (colored blue) in the main pulmonary artery and a mosaic jet along the aortopulmonary connection (12). This turbulent jet was the result of patent ductus arteriosus flow, which was best obtained at the distal ductal orifice of 1.4 cm in diameter (Figs. 4A and 4B).

Cardiac catheterization revealed a net bidirectional shunt at the ventricular level and arterial O_2 desaturation of 85%, with peak ventricular systolic pressure approximating aortic systolic pressure. Due to congenital anatomic alterations, the catheter could not enter the pulmonary artery. Right ventricular cineangiocardiographic examination revealed simultaneous filling of the aorta and main pulmonary artery. Aortography revealed simultaneous filling of the aorta and both pulmonary arteries at the thoracic level, with catheter-detected mean pulmonary artery pressure of 32 mm Hg. Echocardiographic findings correlated well with the cardiac catheterization findings. Coronary arteriography revealed normal coronary arteries.

A curiosity in this patient was a coincidental quadricuspid aortic valve (13,14), with a fourth leaflet between the right and left coronary cusps, allowing for an echocardiographic X-shaped pattern of closed position of the valve in a short axis view (Fig. 5).

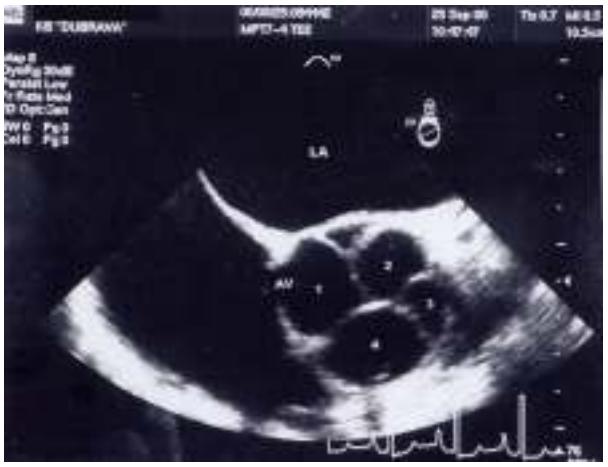

Figure 5. Patient with tetralogy of Fallot and patetent ductus arteriosus. Transesophageal short axis view of the quadricuspid aortic valve (AV) in diastolic closed position, with a characteristic "X" configuration of the cusps (1-4). LA – left atrium.

Discussion

An unoperated patient with tetralogy of Fallot has become a rarity in developed countries because almost all patients undergo palliation or repair in childhood. The reason for avoiding operation of the anomaly in our patient was probably poor motivation of the patient's parents during his childhood because of the absence of symptoms.

In a series of 403 patients aged from 3 months to 41 years, who underwent intracardiac repair of tetralogy of Fallot, the major associated anomalies included atrial septal defect in 24%, pulmonary atresia in 7%, left superior vena cava in 4.5%, and patent ductus arteriosus in 2.5% of the patients (15). In our case, significant narrowing occurred at the level of the pulmonary infundibulum and annulus, which were narrower than the pulmonary trunk and could be visualized in transesophageal view but not in parasternal short axis and subcostal view because of disturbed and complex geometry of the region (8). As the survival of an unoperated patient is determined by the severity of obstruction of the right ventricular outflow tract and pulmonary vasculature (7), mild infundibular and valvular stenosis in our patient with acyanotic adult tetralogy of Fallot indicated a less pronounced resistance to right ventricular outflow, with consequent moderate right ventricular hypertrophy.

The ventricular septal defect was almost as large as the aortic valve and Doppler echocardiography revealed right ventricular peak systolic pressure essentially equal to aortic systolic pressure. These two phenomena implicated simultaneous filling of the aorta and pulmonary artery. As mild pulmonary obstruction causes right-to-left shunt, ventricular septal defect by itself causes left-to-right shunt, resulting in a hemodynamically negligible bidirectional shunt. Large patent ductus arteriosus supplied with blood from both ventricles through a large overriding aorta provides an alternate pathway for blood to reach the lungs, creating pulmonary capillary flow that greatly exceeds pulmonary arterial flow. The flow across the patent ductus arteriosus went from the left (aorta) to the right (pulmonary artery) shunt, with pulmonary circulation returning to the left ventricle and systemic circulation returning to the right ventricle at diastole. Owing to these hemodynamic alternations in our acyanotic case of adult form of tetralogy of Fallot, pulmonary blood flow should probably be twice the systemic flow, with normal arterial oxygen saturation. Persistent ductus arteriosus and the presence of compensatory arteriovenous communications produced a continuous flow load on the left ventricle, which resulted in moderate left ventricular hypertrophy, yet without the symptoms of pulmonary congestion or cardiac decompensation. Right ventricular failure was prevented by the ability to "decompress" into the left ventricle.

On the basis of the clinical presentation and diagnostic findings, the possible mechanism of syncope in our patient could be a shunt worsening on physical effort. If we presume that systemic resistance decreased with physical effort of the patient, the magnitude of the flow through the right-to-left shunt might have increased, since the right ventricular outflow tract obstruction was fixed. This shunting could result in significant arterial hypoxia, which in turn might have precipitated a syncopal episode.

Intracardiac repair of tetralogy of Fallot can be performed with a reasonable risk at any age. However, most authors' experiences suggest the optimal age for elective surgery to be between 6 and 10 years (16). No case of surgical palliation or total correction of adult tetralogy of Fallot with patent ductus arteriosus has been reported in the last 40 years and none with additional quadricuspid aortic valve. Only two

cases of adult tetralogy of Fallot effectively saturated by a patent ductus arteriosus as a natural palliative event have been described, one with dextrocardia, bronchiectasis, and pulmonary tuberculosis at the age of 24 and another at the age of 77 years (17,18). In the latter case, however, the diagnosis was made postmortem. In the sense of these empirical and clinical aspects, our patient was advised to undergo surgical operation, which would involve closure of ventricular septal defect with a patch and patent ductus arteriosus ligation and suture closure as well as possible relief of the right ventricular outflow tract. The operative mortality of tetralogy of Fallot is less than 5% in children and it can be significantly higher in adults (19-21). Our patient has not yet accepted surgical operation.

In conclusion, the life history and clinical manifestations in this patient with tetralogy of Fallot and patent ductus arteriosus, who has reached adulthood without the benefit of surgical palliation or total correction, suggest that life expectancy primarily depends on the degree of pulmonary stenosis, patent ductus arteriosus, and an early development of collateral circulation to the lungs. These natural aortopulmonary anastomoses were efficient in allowing adequate pulmonary blood flow, functioning as a surgical palliative aortopulmonary shunt.

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