LATE DISCOVERY: UNCOVERING TETRALOGY OF FALLOT IN ADULTHOOD- A CASE REPORT

Abstract

Tetralogy of Fallot (ToF) is considered the most frequent cyanotic congenital heart abnormality with a low adulthood survival rate if kept untreated. The majority of cases are symptomatic during infancy and mandate early treatment. Few instances of survival to asymptomatic middle-age patients have been reported, and they are decreasing due to early detection. We report the case of a 44-year-old trisomic male, with poor socioeconomic conditions, with an illness-free past medical history presented for preoperative cardiac assessment for cholecystistis. Tetralogy of Fallot was fortuitously discovered and the patient underwent total correction of ToF. In the immediate post-operative period; the patient presented a complete atrioventricular block, for which a double chamber pacemaker was implanted. Echocardiographic follow-up showed no residual shunt, no pulmonary insufficiency. Our patient had an excellent post-operative and six-months follow-up profile. Thorough physical examination of newborns and a screening echo in the early life may aid in detecting the disease earlier.

Keywords: cholecystistis, Tetralogy of Fallot, dyspnea, blood pressure

Introduction:

Being a frequent cyanotic congenital heart disease, Tetralogy of Fallot (ToF) is often diagnosed in early childhood. In fact, it is associated with a low survival rate if not treated at an early stage. Instances of survival to asymptomatic middle age are rare and decreasing due to improvements in early detection. In this case, we present an adult who lived his live without symptoms until a recent diagnosis of ToF. The patient successfully underwent surgery with satisfying results. This case highlights the potential to diagnose such condition in older individuals, even with advancements of medical screening and treatment.

Case presentation

A 44-year-old trisomic male with an illness-free past medical history presented for preoperative cardiac assessment for cholecystistis. With a personal history of gout disease under treatment. The patient was doing well throughout his life, and has never presented with anoxic crisis. The patient was fairly asymptomatic of his life, complained only of dyspnea on exertion, and his vitals were normal apart from an approximately blood pressure of 150/90 mmHg in all limbs. Physical examination, the patient was pink, a revealed an elevated jugular venous pressure and a harsh systolic ejection murmur best heard at the left upper sternal border, a blood saturation of 93%. Basic blood tests were normal; no polycythemia, however, ECG exhibited a right bundle branch block (RBBB). (Figure 1)

An urgent echocardiogram for the patient showed a unique cono ventricular septal defect (VSD) with a right to left shunt (Figure 2A), severe pulmonary stenosis (Figure 2B), overriding aorta (Figure 2C) and hypertrophied right ventricle (Figure 2D).Otherwise, pulmonary venous return was normal and there was no abnormality in left cavities. After establishing the diagnosis of ToF regular, the patient was referred to cardiac surgery for a repair. A chest scan was requested as part of the pre-operative assessment and showed few aorto-pulmonary collaterality (Figure 3)

Total surgical correction

Total correction of ToF was executed by infundibular approach with closure of the VSD by a pericardium patch, resection of the septal and parietal band, associated with valvotomy by widening of the pulmonary tract through a bougie.

In the immediate post-operative period; the patient presented a complete atrioventricular block, for which a double chamber pacemaker was implanted. Echocardiographic follow-up showed no residual shunt, no pulmonary insufficiency.

Six month later, the patient was doing well, complained of no symptoms and rhythmically electro-trained.

Discussion:

In the present era, Tetralogy of Fallot (TOF) is typically diagnosed during childhood. However, in comparison to 50 years ago, a review of ToF cases revealed that 17, 6% of the studied population were more than 25 years old at the time of diagnostic [1] .Advances in medical knowledge and practice have led to earlier diagnosis and treatment, but for patients with benign clinical presentation and limited access to healthcare; diagnosis remains delayed .In fact, our patient didn't had any troubling symptoms.

Even though uncorrected ToF survival is uncommon, it has been reported that around 10% of affected persons can survive to adulthood, and only 5% reach 40 years of age[2,3] .The survival of our patient and the diagnosis of his condition at the age of 44 years old make this case exceptional. Moreover, similar to the different degrees of presenting symptoms due to anatomical variants, unrepaired ToF might be diagnosed late as survivors could have favorable anatomy that generally permits better pulmonary flow, in contrast to those who presented earlier in their life [4].Mechanisms that explain longevity in patients who remained undiagnosed and survived to their adulthood may be attributed to having a protective pulmonary stenosis and a slow development of subpulmonary obstruction, left ventricular hypertrophy, and extracardiac shunting or systemic to pulmonary shunt [5]. In our case, the reason for delayed presentation is not completely clear, but it may be attributed to slow development of pulmonary valve stenosis which explained the approximately correct pulse oxygen saturation.

Cardiac catheterization is essential in managing ToF in adults as it not only anatomically characterizes the pulmonary arteries but also identifies unexpected anomalies like aorto-pulmonary collateral arteries, which are present in 15 % of ToF patients.[6] Addressing such abnormalities through restorative procedures can streamline surgical management. Our patient did not undergo cardiac catheterization, but a CT angiography allowed for the non-invasive characterization of the anatomy of the pulmonary arteries and their branches.

A delayed diagnostic can lead to a postpone treatment. While patients undergoing ToF repair generally achieve excellent outcomes [7], surgical management often results in anatomical and functional complications, most

commonly valve regurgitation and right ventricular dysfunction.Prolonged chronic pulmonary valve regurgitation can lead to right ventricular dilated and failure, increased triscupid regurgitation, diminished exercise performance, and supraventricular or ventricular arrhythmias [8]. In our case, total correction of ToF was executed by infundibular approach with closure of the VSD by a pericardium patch, resection of the septal and parietal band, associated with widening of the pulmonary tract through a bougie.

Greater mortality in the pediatric group is attributed to prolonged right ventricular dysfunction and underdevelopment of the pulmonary artery, leading to chronic cyanosis and a diminished quality of life [9,10]. The degree of pulmonary stenosis played a protective role in our case, allowing for viable oxygenation and the patient's survival up to this age.

Patients require lifelong follow-up after repair, as mostly will eventually need pulmonary valve replacement. The management of asymptomatic patients largely depends on their age, and physiological evaluations should be conducted in those at high risk of arrythmia complications prior to any reintervention [11]. Echocardiographic follow-up showed no residual shunt, no pulmonary insufficiency; ruling out any need of revalvulation so far. The patient had a well-functioning valve and stable right ventricular function that can hopefully reduce the likelihood of needing pulmonary revalvation.

Delayed intervention impacts more than just physical health. Adults with repaired ToF frequently face considerable mental health challenges, chich not only disrupt their daily life but also significantly impact their treatment plans. [10].Psychosocial support is therefore required.

Conclusion:

We presented a case of a patient who was diagnosed to have ToF during his adulthood. The case postulates that patients with primary congenital diseases may remain undiagnosed until an older age despite medical advances. Delaying the diagnosis and management of ToF cases increases the risk of adverse outcomes. However, our patient had an excellent post-operative and six-months

follow-up profile. Thorough physical examination of newborns and a screening echo in the early life may aid in detecting the disease earlier.

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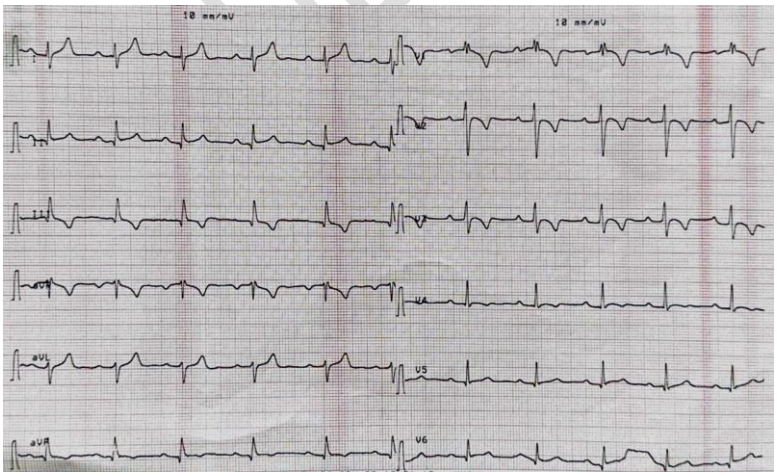

Figure 1: EKG of the patient showing right bundle branch block with negative T waves in the anterior and inferior leads.

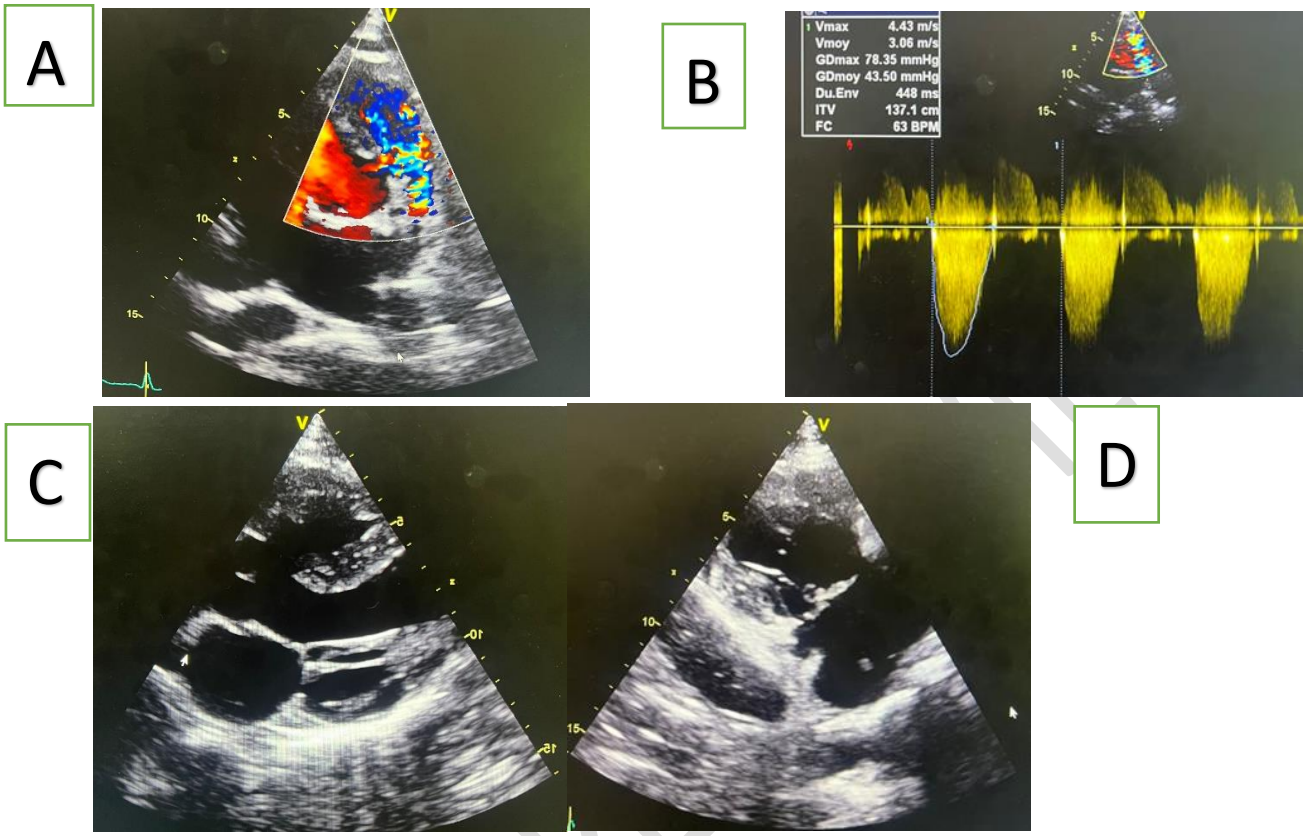

Figure 2:

Patient‘s echocardiogram showing: ventricular septal defect with a right to left shunt (A), pulmonary stenosis (B), overriding aorta (C), severe right ventricular hypertrophy(D)

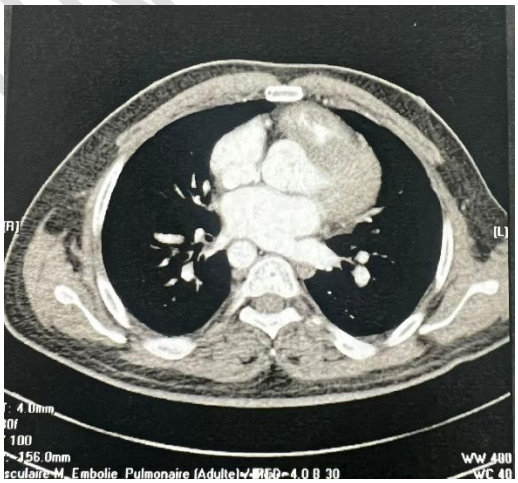

Figure 3: A chest scan was requested as part of the pre-operative assessment with presence of aorto-pulmonary collaterality