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Case Report

An exceptional survival in an unoperated tetralogy of Fallot in a 66-year-old man: A case report[☆]

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ARTICLE INFO

Article history:

Received 21 August 2022

Revised 15 September 2022

Accepted 18 September 2022

Keywords:

Tetralogy of Fallot

Congenital heart disease

Cyanosis

management

Unoperated

ABSTRACT

Tetralogy of Fallot is the most common cyanotic congenital heart disease in children which comprises an overriding aorta, right ventricular outflow obstruction, ventricular septal defect, and right ventricular hypertrophy. It has an elevated early mortality rate without surgical correction, with most patients dying in childhood. Only 2% of patients survive past the age of 40 years without surgical intervention. Very few cases of survival to middle age have been reported, particularly after the fourth decade. In this article, we present a case of a 66-year-old male with an unoperated tetralogy of Fallot, which is one of the longest time periods of diagnosis. Despite tetralogy and having right ventricular dysfunction, this patient presented with fatigue, exertional dyspnea, cyanosis, and systemic hypertension. Considering the patient's comorbid conditions and the risk associated with the surgery, the patient was managed conservatively. To our knowledge, this is the oldest unoperated tetralogy of Fallot case reported in Nepal.

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Introduction

Tetralogy of Fallot (TOF) is the most common congenital cyanotic heart defect in childhood but is rarely seen in adulthood. An estimated 400 live births per million are affected by

this condition [1]. Coexisting features of the disorder include the overriding aorta, ventricular septal defect (VSD), and obstructed right ventricular outflow tract (RVOT) [2]. Most patients would die during their childhood without surgical repair. If left untreated, mortality after age 40 is 95%. Historically, 66% of persons with TOF who were not surgically treated lived

[☆] Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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<https://doi.org/10.1016/j.radcr.2022.09.062>

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to be 1, 49% lived to be 3, and 24% lived to be 10 [3]. Over the fourth decade, survival in TOF is only 2%, and survival past the sixth decade is very unlikely [4,5].

Here, we present a case of a 66-year-old man with an uncorrected TOF with fatigue, dyspnea, cyanosis, and systemic hypertension. Considering the patient's comorbid conditions and risk associated with the surgery and complying with the patient's desire to go for conservative therapy, we decided to manage medically. To our knowledge, this is the oldest unoperated TOF case reported in Nepal.

Case report

A 66-year-old male presented at our hospital complaining of fatigue, swelling in the legs, and exertional dyspnea in the past 4 years. On clinical examination, cyanosis and edema were seen in both lower limbs. He also described experiencing exertional dyspnea and shortness of breath intermittently. Due to this, he was not able to work for a prolonged period without rest. His activities were limited to simple work and he could not do strenuous activities. Renal function test revealed a creatinine level of 132 $\mu\text{mol/L}$ (normal: 60–110 $\mu\text{mol/L}$). He was aware of his unrepaired congenital heart defect. Previously, he refused to undergo surgical repair at 25 years old because of the risk involved. Oxygen saturation was 85% at rest, and blood pressure was 165/90 mm Hg. On further workup, hemoglobin was 16 g/dL.

On auscultation over the precordium, a systolic ejection murmur was heard in the left second intercostal space with a right ventricular heave and palpable thrill. Electrocardiogram (ECG) was done, which showed sinus rhythm, prominent P-wave in V1, right-axis deviation, 9 mm R wave in V1, and R/S ratio of 1.5, that is, right ventricular hypertrophy. Two-dimensional transthoracic echocardiography showed right ventricular hypertrophy, VSD, overriding of the aorta over the septum, and aorto-mitral continuity (Fig. 1), confirming TOF.

The patient refused to undergo surgical correction due to his advanced age and associated comorbidities like impaired renal function. The patient was informed of the surgical risk as well as conflicting data regarding long-term prognosis and risk of sudden cardiac death. He was treated conservatively with diuretics (Furosemide) and angiotensin converting enzyme (ACE) inhibitor (Captopril) for hypertension. The patient's condition significantly improved and was discharged. On follow-up at 3 months, the patient symptoms significantly improved, and hypertension was in control.

Discussion

The most common cyanotic congenital heart defect is the TOF. Asymptomatic patients with undiagnosed TOF may mistake it for a small VSD. Heart failure or cyanosis is usually diagnosed in newborns or in the first few years of life. Several factors contribute to this disease, but reported associations include retinoic acid intake, phenylketonuria, untreated maternal diabetes, and chromosomal abnormalities (trisomies 18, 21 and

microdeletions of chromosome 22). Families are at risk for recurrence by 3% [6].

If left untreated, mortality after age 40 is 95%. Historically, 66% of people with TOF who were not surgically treated lived to be 1, 49% lived to be 3, and 24% lived to be 10 [3]. Without intervention, fewer than 2% of patients reach the age of 40 [4]. Twelve years is the median unoperated survival rate. Typically, the pathophysiology depends on the severity of RVOT obstruction, which is accompanied by right-to-left shunting that is dictated by systemic vascular resistance. This rare occurrence occurs in patients with infundibular pulmonary stenosis such that hemodynamics are balanced, that is, the patient does not suffer from severe pulmonary hypertension (Eisenmenger's) from a large left-to-right shunt, but maintains enough pulmonary blood flow to prevent cyanosis. There are some patients with heart failure symptoms when the left-to-right shunt is dominant. Cyanosis occurs as right-to-left shunting across the VSD increases as the RVOT obstruction increases.

According to previous studies, unoperated survivors share 3 common characteristics: left ventricular hypertrophy (LVH), hypoplastic pulmonary artery with the moderately slow development of subpulmonary obstruction, and systemic-pulmonary artery collaterals [7]. Our patient had LVH, overriding of the aorta and aorto-mitral continuity, similar to other published cases. The development of LVH is presumed to delay right to left shunting and often happens later in adulthood [7]. There were no systemic-pulmonary artery collaterals in our patient.

Children and infants usually undergo repair during their formative years. The ideal age for repair is a matter of debate. Surgical correction of TOF is performed during infancy, aiming for total correction by 9 months of age [8]. Infants as young as 1 year of age are often operated on, and perioperative mortality is less than 5%. In transatrial/transpulmonary surgery, the VSD is closed, the right ventricular muscle is resected or infundibulotomized, and the pulmonary arteries are enlarged as needed. Patients who have surgery have excellent long-term outcomes, and most lead active and productive lives, while those without surgery have a poor life expectancy [9]. Throughout the course of follow-up, sudden cardiac death due to malignant ventricular arrhythmias originating from the ventricular septal patch or from the site of a surgical scar can occur and lead to reduced survival [10].

In untreated cases of TOF, right ventricular hypertrophy leads to right ventricular failure, which quickly progresses to biventricular failure [11]. Some patients, however, survive without treatment. It is rare for the clinician to encounter an adult with an unrepaired TOF. Among patients with severe obstructions not treated surgically, 25% die within the first year, 40% within 3 years, 70% within 10 years, and 95% within 40 years. Patients who die without surgical treatment most commonly suffer from hypoxic spells (62%), cerebrovascular accidents (17%), and brain abscesses (13%) [12]. Almost no case reports have documented late survival without surgery, and this is usually due to less severe RVOT obstruction and right-to-left shunting, which becomes evident during exertion [3]. After 20 years, the reported survival rate for patients with TOF older than 10 years is between 10% and 12%, and for those over 30 years, the survival rate is around 6% [13].

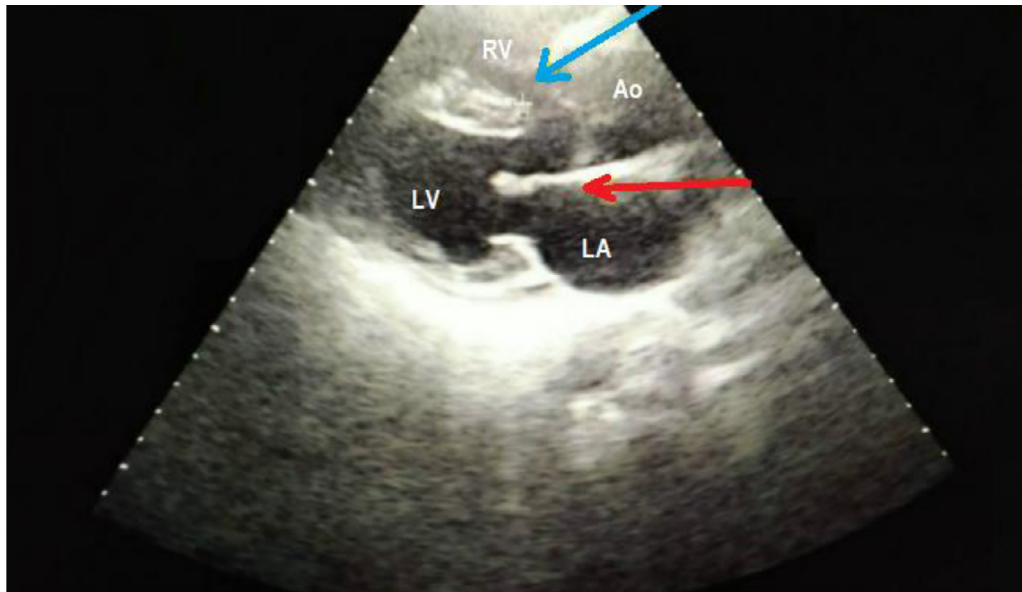


Fig. 1 – Two-dimensional transthoracic echocardiogram demonstrated characteristics of tetralogy of Fallot: ventricular septal defect (VSD) and the overriding aorta over the septum (blue arrow), aorto-mitral continuity (red arrow), ventricular septal defect, and right ventricle (RV) hypertrophy. Ao, aorta; RV, right ventricle; LA, left atrium; LV, left ventricle.

As a result of their frail myocardium, bleeding from pulmonary collateral circulation, and other complications such as brain abscess and endocarditis, the elderly with TOF are said to have a higher operative risk than younger patients. Complete repair of TOF is possible in patients over the age of 40, but the risks of surgery are high. In patients who underwent total correction at the ages of 40–60 years, operative mortality was found as 3%, whereas survival was 92% at 5 years and 74% at 10 years [13]. Drugs such as beta-blockers and ACE inhibitors may be used in these patients. Even though the overall survival is reduced after successful surgery, the role of surgical management in elderly patients is controversial. There is a poor long-term prognosis, and the risk of sudden cardiac death is present mainly in the right ventricle [10]. In our case, we decided to manage medically complying with the wish of the patient, and also because there is inadequate data to support surgery at this age, especially in the presence of overt RV dysfunction.

Conclusion

In extremely rare cases, some patients with TOF can reach older ages without surgery. It is important for careful echocardiographic analysis to diagnose this condition. The role of operative management in elderly patients is controversial as the overall survival is significantly reduced even after surgery. In the event of a refusal of surgery or poor candidates for surgery, medical management is required. Drugs such as ACE inhibitors, diuretics, and beta-blocker may be tried.

Patient consent

Written informed consent for the publication of this case report was obtained from the patient.

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