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Late Presentation of Tetralogy of Fallot and Uncommon Survival in A 31-Year-Old Patient

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Abstract

Background: Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease encountered in clinical practice. It consists of ventricular septal defect (VSD), overriding aorta, right ventricular outflow tract obstruction (RVOTO), and right ventricular hypertrophy (RVH). Clinical severity depends on the degree of RVOTO, presence or absence of extracardiac shunts or collaterals, and other factors. Only 10% of untreated patients survive into adulthood.

Objective: To describe late presentation of TOF and uncommon survival in a 31-year-old patient.

Case Report: A 31-year-old male presented at the cardiology clinic of a secondary healthcare facility in Gombe, Gombe State, Nigeria, with complaints of exercise intolerance. His symptoms started at 4 years of age, and he did not have any significant limitation in day-to-day activities. He had no history of prior cardiac investigation or care. Significant examination findings were plethora, central cyanosis, grade 4 finger clubbing, height 170 cm, weight 70 kg, body mass index 24.2 kg/m^2, grade 3/6 systolic murmur at the left upper sternal border, and blood pressure of 110/70 mmHg. Packed cell volume was 71%, SPO_2 was 84%; chest X-ray showed a normal-sized heart with boot-shaped appearance, presence of pulmonary bay, and lung oligemia. Electrocardiographic findings were in keeping with RVH. Transthoracic echocardiography revealed a large subaortic VSD, overriding aorta, severe valvular pulmonary stenosis, and right ventricular hypertrophy.

Conclusion: There should be a high index of suspicion for CHDs in adults presenting with dyspnea on exertion and cyanosis. Also, there is a need for early diagnosis of CHD through prompt referral of patients with respiratory or cardiac symptoms to centers that can perform echocardiography.

Keywords

Tetralogy of Fallot, Late Presentation, Survival

Background

Tetralogy of Fallot (TOF) is the most commonly encountered cyanotic congenital heart disease (CHD) in individuals beyond infancy. This complex cardiac

anomaly is characterized by a quartet of specific structural defects: a ventricular septal defect (VSD), an overriding aorta, right ventricular outflow tract obstruction (RVOTO), and right ventricular hypertrophy (RVH). The combined physiological effects of these defects lead to a right-to-left shunting of deoxygenated blood, resulting in systemic cyanosis [1,2].

The clinical manifestations and severity of TOF are profoundly influenced by the degree of RVOTO. Patients with more severe obstruction typically exhibit profound cyanosis, while those with milder forms may present with less overt cyanosis or even appear "pink." The presence or absence of extracardiac shunts or collateral circulation also plays a crucial role in determining the patient's clinical status and long-term prognosis. In rare instances, patients with mild RVOTO may present with symptoms more akin to congestive heart failure rather than cyanosis, a phenomenon referred to as "pink TOF."

Historically, the prognosis for untreated TOF was dismal, with only a small fraction of patients surviving into adulthood. Longitudinal studies indicate that approximately 10% of individuals with unrepaired TOF may survive beyond their 20s, [3] with survival rates drastically declining thereafter. This grim prognosis underscores the imperative for early diagnosis and timely surgical intervention. With advancements in diagnostic modalities, particularly the widespread availability of transthoracic echocardiography, most cases of TOF are now identified early in life, often within the first few months. Current guidelines advocate for surgical repair between 3 and 11 months of age to optimize outcomes and prevent long-term complications [4].

However, in resource-limited settings or due to various socio-economic factors such as financial constraints, lack of specialized facilities, or insufficient expertise, many patients may not receive definitive surgical correction within the recommended timeframe [5]. Consequently, these individuals may present with late-stage disease or endure prolonged periods with uncorrected defects, posing significant clinical challenges.

The natural history of unrepaired TOF is marked by complications, including erythrocytosis, paradoxical emboli, brain abscesses, infective endocarditis, and arrhythmias [3]. These complications contribute significantly to the high mortality rates observed in untreated populations. Therefore, understanding the unique physiological adaptations and risk factors associated with late presentation of TOF is crucial for clinicians, particularly in regions where access to advanced cardiac care remains limited.

Objective

The primary objective of this case report is to present a detailed account of a patient diagnosed with Tetralogy of Fallot who exhibited an unusually late clinical presentation and remarkable survival into his fourth decade without surgical intervention. This case aims to contribute to the limited body of literature on the natural history of uncorrected TOF in adult populations.

Methodology

The clinical records pertinent to the patient's diagnosis, presentation, and subsequent management were retrieved from the hospital's comprehensive medical archives. This process adhered strictly to institutional protocols for data access and patient confidentiality. Prior to the commencement of this publication, fully informed consent was obtained from the patient. This consent encompassed the use of his de-identified clinical data for educational and research purposes, ensuring ethical compliance and patient autonomy.

Clinical, laboratory, electrocardiographic, and echocardiographic findings were systematically reviewed and analyzed to provide a comprehensive understanding of the patient's condition and the physiological adaptations enabling his prolonged survival.

Case Report

A 31-year-old male presented to the cardiology clinic of a secondary healthcare facility in Gombe, Northeast Nigeria, with his primary complaint being progressive exercise intolerance. This symptom had an insidious onset at four years of age. Remarkably, despite the chronic nature of his symptoms, he reported no

significant limitations in his day-to-day activities until recently, indicating a prolonged period of physiological adaptation to his condition. Notably, there was no history of prior cardiac investigations, consultations, or any form of cardiac care, suggesting a lifelong journey with undiagnosed Tetralogy of Fallot.

Upon physical examination, several significant findings were noted. The patient exhibited pronounced plethora (an excess of blood, leading to a ruddy complexion) and central cyanosis, indicative of chronic hypoxemia. Grade 4 finger clubbing (**Fig-1**), a classic sign of long-standing hypoxemia, was evident. Anthropometric measurements revealed a height of 170 cm, weight of 70 kg, and a Body Mass Index (BMI) of 24.2 kg/m^2, all within normal ranges. Auscultation of the precordium revealed a grade 3/6 systolic murmur at the left upper sternal border, consistent with significant outflow tract obstruction. His blood pressure was 110/70 mmHg.

Fig-1: Grade 4 Finger Clubbing

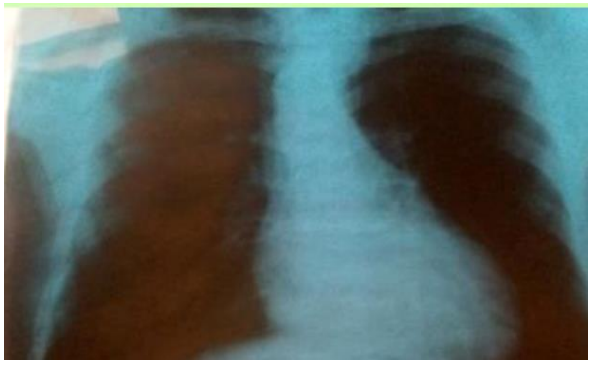

Fig-2:

Chest X-Ray Showing Boot-Shaped Heart, Pulmonary Bay and Pulmonary Oligaemia

Laboratory investigations showed a packed cell volume (PCV) of 71%, indicating severe secondary polycythemia, a compensatory mechanism for chronic hypoxemia. Peripheral oxygen saturation (SPO_2) was measured at 84%, further confirming significant cyanosis. A chest X-ray revealed a normal-sized cardiac silhouette with a characteristic "boot-shaped" appearance (coeur en sabot), alongside the presence of a pulmonary bay and lung oligemia (**Fig-2**), all classic radiological features of Tetralogy of Fallot. Electrocardiographic (ECG) findings were consistent with prominent right ventricular hypertrophy (RVH).

The definitive diagnosis was established through transthoracic echocardiography, which provided comprehensive structural and functional details. It unequivocally revealed a large subaortic ventricular septal defect (VSD), an overriding aorta, severe valvular pulmonary stenosis (the primary determinant of clinical severity), and significant right ventricular hypertrophy (**Fig-3** and **Fig-4**). These findings collectively confirmed the diagnosis of Tetralogy of Fallot.

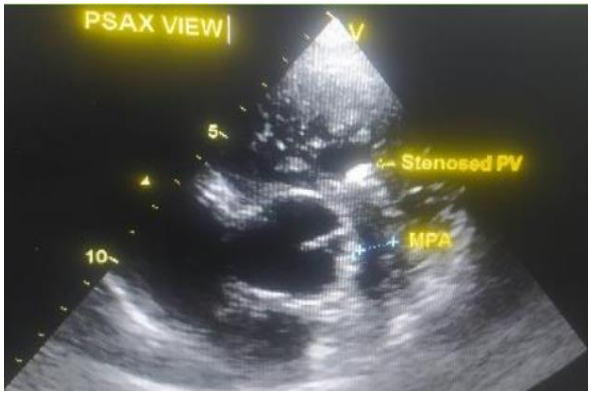

Fig-3:

Transthoracic Echocardiography in Parasternal Short Axis View Showing Valvular Pulmonary Stenosis

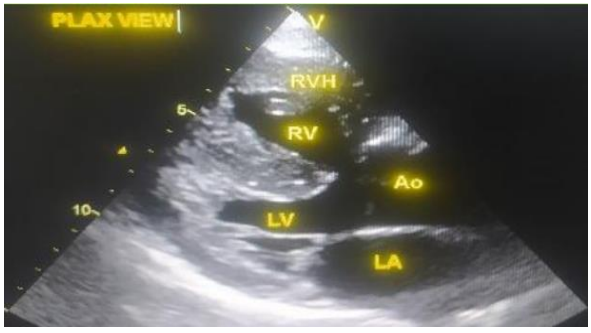

Fig-4:

Transthoracic Echocardiography in Parasternal Long Axis View Showing a Large Ventricular Septal Defect, Overriding Aorta and Right Ventricular Hypertrophy

Given the patient's late presentation and unique survival, the immediate management focused on

symptomatic relief and stabilization. The patient was counseled regarding the nature of his condition and the potential for surgical intervention. However, due to the advanced age of presentation and the chronic nature of his cardiac remodeling, a multidisciplinary discussion was initiated to assess the feasibility and risks associated with corrective surgery. The current plan involves careful monitoring, medical management of his symptoms, and a thorough pre-operative assessment to determine if he is a suitable candidate for future surgical repair, acknowledging the limited surgical facilities and expertise available in his region for such complex adult congenital heart disease cases.

Discussion

Tetralogy of Fallot (TOF) is a complex congenital heart defect characterized by four key features: ventricular septal defect, pulmonary stenosis, right ventricular hypertrophy, and an overriding aorta. It may also be associated with atrial septal defect (ASD), in a case of Pentalogy of Fallot. The basic pathology of TOF is due to the underdevelopment of the right ventricular infundibulum, which results in an anterior-cephalad deviation of the infundibular septum [2]. This malalignment determines the degree of right ventricular outflow tract obstruction (RVOTO), which in turn is the primary determinant of the clinical phenotype and severity of cyanosis. A recent case report highlights the importance of prompt diagnosis and surgical intervention in managing this condition [6].

The late presentation of TOF in a 31-year-old patient, as described in this case, represents an exceedingly uncommon survival scenario. The vast majority of individuals with uncorrected TOF succumb to complications such as severe hypoxemia, cerebral abscesses, infective endocarditis, and paradoxical embolism before reaching adulthood [7,8]. The remarkable longevity observed in this patient suggests a unique set of adaptive physiological mechanisms, potentially involving a delicate balance of the four defects that permitted adequate systemic perfusion and oxygen delivery despite significant underlying pathology. The development of systemic-to-pulmonary collateral arteries might have contributed to his prolonged survival [9].

The age at diagnosis for this patient is later than that of many reported patients with late presentation of TOF. The late presentation might be due to poor health-seeking behavior. Survival till age 31 years might be due to the presence of aortopulmonary collaterals, which would improve pulmonary blood flow. Other studies have also reported survival of patients with unrepaired TOF into the 6th to 9th decades of life; some factors attributed to survival include systemic hypertension and milder degrees of RVOTO [10-12]. In these patients, however, the diagnoses of TOF were made before 20 years of age in some, and in the early and late 20s in others. One patient was diagnosed at the age of 31 years in 1961, an era when echocardiography was just being discovered [12].

A report by Gorla et al. [10] highlighted the survival of an 85-year-old man with unrepaired TOF, an even more exceptional case that underscores the extreme variability in the natural history of this condition. Similarly, Makaryus et al. [11] reported on a patient surviving to age 52 years with unrepaired TOF. These rare cases provide invaluable insights into the physiological adaptations that can occur in the absence of surgical correction, challenging the conventional understanding of the condition's absolute lethality without intervention. Alkashkari et al. [13] reported a case of a 29-year-old man who was diagnosed with TOF for the first time at that age; he underwent successful surgery, highlighting the possibility of successful late repair.

However, it is crucial to emphasize that such prolonged survival without intervention is atypical and should not diminish the importance of early diagnosis and prompt surgical repair. Surgical correction of TOF, ideally performed in infancy, dramatically improves prognosis, prevents long-term complications, and enhances quality of life [14]. The challenges faced by adult survivors of unrepaired TOF - including right ventricular dysfunction, arrhythmias, and sudden cardiac death - necessitate continuous, specialized cardiological follow-up and consideration for late corrective or palliative interventions, even if the optimal window for surgery has passed. This case reinforces the critical need for increased awareness among clinicians regarding the possibility of adult presentation of CHDs, particularly in regions with limited healthcare access.

Limitations

Unavailability of Cardiac CT Angiography:

A significant limitation in the diagnostic work-up of this patient was the unavailability of cardiac Computed Tomography Angiography (CCTA) at the secondary healthcare facility where he was evaluated. CCTA would have been immensely beneficial in providing a highly detailed three-dimensional anatomical assessment of the great arteries and branch pulmonary arteries. Such detailed imaging could have precisely delineated the extent of pulmonary artery hypoplasia or stenosis and identified any additional systemic-to-pulmonary collateral arteries, which are often crucial compensatory mechanisms in uncorrected cyanotic heart diseases. The absence of this advanced imaging modality limited a complete understanding of the patient's complex vascular anatomy and collateral circulation, which are vital for surgical planning and prognostication.

Lack of Long-Term Follow-up Data:

This report captures the patient's condition at the point of presentation. Longitudinal follow-up data, including serial echocardiographic assessments and functional capacity evaluations, would provide deeper insights into the progression of his cardiac remodeling and the long-term effectiveness of any initiated medical management. Without this, the complete natural history of his specific case remains partially obscured beyond the initial assessment.

Despite these limitations, the case provides significant contributions to the understanding of adult TOF and emphasizes the diagnostic challenges in resource-constrained environments.

Conclusion

This case report highlights the critical importance of maintaining a high index of suspicion for congenital heart diseases (CHDs) in adult patients, particularly those presenting with symptoms such as exercise intolerance and cyanosis. The remarkable survival of a 31-year-old male with unrepaired Tetralogy of Fallot underscores the adaptive capabilities of the human body, yet also points to significant gaps in early diagnosis and access to specialized cardiac care, especially in developing regions. Timely and accurate diagnosis through prompt referral for echocardiography remains paramount for improving patient outcomes and preventing the life-threatening complications associated with uncorrected CHDs.

Conflict of Interest

The authors have read and approved the final version of the manuscript. The authors declare no conflicts of interest.

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