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AN UNUSUAL ASYMPTOMATIC PRESENTATION OF TETRALOGY OF FALLOT IN ADULTHOOD: A CASE REPORT

Cardiovascular Surgery

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ABSTRACT

Tetralogy of Fallot is most common cyanotic congenital heart disease. Less than 5 % patients reach the age of 40 without surgical intervention. It is very rarely diagnosed in adulthood, striking feature about this case being its asymptomatic nature till 52 years of age. We present a case report of 52 year male suffering from inguinal hernia, during preop investigation for the same, ECG showed atrial flutter, 2D Echo diagnosed TOF and hence referred to our institute for further management. After detailed routine preoperative and electrophysiological workup, operated for intracardiac repair with CABG with permanent pacemaker implantation (DDDR). The postoperative course was uneventful. This case will an addition to very small cohort of untreated adult TOF patients.

KEYWORDS

INTRODUCTION

Tetralogy of Fallot (TOF) was first described by Etienne-Louis Fallot in 1888[1]. It is characterised by four distinct anatomic abnormalities: right ventricular hypertrophy, ventricular septal defect, obstruction of right ventricular outflow tract and overriding of aorta. Early primary repair is the treatment of choice[2]. Delaying the diagnosis and late intervention are associated with poor outcomes[3]. However, as an exception to previous statement, our current case stands out with its asymptomatic course. According to large observational studies, 24% individuals with uncorrected TOF die before the age of 10, only 4% survive beyond their thirties[4,5]. As per another study, only 66% of patients survive up to 1 year of age without surgery, 49% to 3 years and 24% till 10 years of age[6,7]. In this case report, we are presenting a very unique case of adult asymptomatic TOF diagnosed due to presence of atrial flutter on ECG, detailed workup revealing the diagnosis.

Case Report

A 52-year-old asymptomatic male, presented to OPD of our hospital, referred by a general surgeon, with the diagnosis of pulmonary stenosis on echocardiography and ECG showing atrial flutter. This patient was suffering from right inguinal hernia, consulted a general surgeon for the same, revealing arrhythmia in ECG which lead to diagnosis of TOF. No history of congenital heart disease in family.

Physical Examination Findings were as follows:

General Appearance

Average built, Oriented to time, place and person. Not in distress, comfortable on room air and no dyspnea on 6 Feet walk.

Vital Signs Were

Blood pressure 110/60 mmHg, heart rate 53 beats/minute, respiratory rate 18 breaths/minute, temperature 35.8 °C, and oxygen saturation 97% on room air.

General Examination

Clubbing: Grade 3 clubbing present.
Cyanosis: No cyanosis.
Edema: No peripheral edema.

Cardiovascular System (CVS)

S1 and S2 are audible with a pansystolic murmur, graded 3/6 in intensity, was auscultated. The murmur was most prominent along the left parasternal border but was not associated with a palpable thrill.

Abdomen

The abdomen was soft and non-tender on palpation.
Right inguinal hernia detected.

Peripheral Examination

No ankle edema or other signs of peripheral vascular compromise were observed.

Nervous Examination

A brief neurological assessment showed the patient to be alert and fully oriented to person, place, and time. There were no obvious motor or sensory impairments. Examination of cranial nerves II through XII revealed no gross abnormalities. Additionally, there were no clinical signs suggestive of meningeal irritation.

The blood investigations were normal. Chest x-ray showed cardiomegaly with oligemic lung fields, LV type of apex. ECG showed atrial flutter with ventricular rate of 53/min. 2d echo suggestive of small, restrictive sub-aortic VSD with <50% aortic override with bidirectional shunt, severe infundibular PS, hypoplastic pulmonary valve, confluent branch PA's, mild RV dysfunction with normal LV function. CAG suggestive of 90% lesion in LAD, 80% lesion in LCx. Beta-blockers and calcium channel blockers were started in preoperative period but atrial flutter persisted despite these medications. On detailed discussion with patient regarding the management of arrhythmia in post operative period and the possibility of atrial arrhythmia with controlled ventricular rate with occasional episodes of fast ventricular rate, requirement of lifelong anticoagulation for stroke prophylaxis, finally it was decided to opt for an option which provided solution in single setting along with the surgery. Consultation with electrophysiological department also suggested implantation of permanent pacemaker. After a detailed informed consent, patient was planned for surgery. Intraoperatively, PA size was adequate. Sub-aortic VSD was restrictive and dense fibrous tissue responsible for sub-valvular infundibular PS was found. Pulmonary valve was salvageable. LIMA to distal LAD and RSVG to OM major grafts were done. DDDR epicardial pacemaker implantation done. PRV/LV was 0.5 and RV-PA gradient was 15 mmHg. The postoperative course was uneventful. Patient was discharged on postop day 5.

DISCUSSION

Tetralogy of Fallot (TOF) is a congenital cardiac anomaly characterized by a ventricular septal defect, right ventricular outflow tract obstruction, overriding of the aortic root, and right ventricular hypertrophy. It occurs in approximately 3 per 10000 live births, accounting for 7–10% of all congenital heart defects. Clinical presentation typically occurs in the neonatal period, with cyanosis varying in severity based on the degree of right ventricular outflow obstruction. The etiology is multifactorial, with genetic and environmental factors playing a role. Maternal conditions such as diabetes and phenylketonuria, as well as chromosomal anomalies like trisomies 21, 18, and 13, have been associated with TOF, though recent evidence suggests a stronger link with 22q11.2 microdeletion[6].

Survival of TOF patients in adulthood is because of less severe disease and formation of aorto-pulmonary collaterals, which was the case with our patient. The degree of obstruction to pulmonary blood flow determines the clinical state of the patient. Unrepaired TOF presenting beyond the third decade is associated with milder degree of pulmonary obstruction that allows longer survival. Patients with cyanotic heart disease like TOF are known to have dilated large coronary arteries due to chronic hypoxia, traditionally believed to be less prone to develop coronary artery disease[7]. Our patient was an exception from latter point also. The factor which may have led to coronary artery disease in our patient may be chronic smoking, tobacco chewing and family history of CAD. The incidence of hemoptysis and neurological

complications like brain abscess is certainly higher in adults than in children[8]. Also there is high chance of heart failure due to long standing pressure overload and consequent pathological changes on right ventricle[9]. Previously it was believed that old age was incremental risk factor for early mortality following total correction[10], but according to latest case series, old age is no more believed to be a risk factor for increased mortality, due to improved intraoperative and postoperative management[7]. There is not much literature on arrythmia in adult patients of unrepaired TOF, but high prevalence of tacchyarrythmia was found in adult patients of TOF[11]. The prevalence of coronary artery disease in adult TOF patients was 34%, out of which 24% patients underwent CABG[12].

Individuals without surgical intervention are most often killed by hypoxic episodes (68%), cerebrovascular accidents (17%) and brain abscess (13%)[13].

In diagnosing congenital heart diseases like Tetralogy of Fallot (ToF), readily available tools such as electrocardiography (ECG) and echocardiography are indispensable, offering a non-invasive approach. An ECG can reveal right axis deviation and right ventricular hypertrophy, indicative of the strain on the heart caused by ToF. Furthermore, echocardiography (2D ECHO) is crucial for confirming the diagnosis of ToF, as it can visualize the key anatomical defects, including ventricular septal defect (VSD), overriding aorta, and right ventricular outflow tract (RVOT) obstruction[14].

While ECG and standard echocardiography are essential for the initial diagnosis and monitoring of Tetralogy of Fallot (ToF), particularly in resource-limited settings due to their accessibility, advanced imaging modalities are often required for comprehensive anatomical and hemodynamic evaluation. Cardiac MRI (cMRI) is central in assessing ventricular function, pulmonary valve pathology, and right ventricular outflow tract (RVOT) morphology.[15]

Tetralogy of Fallot (TOF) is a cyanotic congenital heart disease associated with high mortality rates among unrepaired patients. Survival beyond 20 years is limited to approximately 10%, with only 3% reaching 40. In contrast, over 90% of patients undergoing surgical repair survive into adulthood. In developed countries, most individuals with TOF receive timely surgical intervention, which alleviates right ventricular outflow tract (RVOT) obstruction and significantly reduces mortality. However, in low-resource settings, access to surgical repair remains limited, particularly among patients from low socioeconomic backgrounds. This disparity contributes to poorer long-term outcomes and increased mortality in these populations[16].

In some cases, patients may not receive an accurate diagnosis during childhood, as was observed in our case. The first clinical suspicion of Tetralogy of Fallot (TOF) only arose when the patient reached adulthood. Confirming the diagnosis required advanced imaging studies, which were often financially inaccessible and frequently unavailable in his setting, both during initial assessment and routine follow-up. The lack of continuous monitoring and access to diagnostic tools contributed significantly to the delayed diagnosis and management of his condition. This case highlights the critical importance of making bedside echocardiography available, even in resource-limited environments. Point-of-care ultrasound can enable earlier detection and timely intervention in congenital heart diseases, ultimately improving patient outcomes.

In our patient, arrythmia namely atrial flutter was the point which lead to diagnosis of TOF. This particular event highlights the importance of arrhythmias, however asymptomatic they may be. The possible mechanism of atrial arrhythmia may be the extensive fibrosis that was present surrounding the VSD.

CONCLUSION

Very few patients make it in their 50's, asymptomatic through their entire life, with the diagnosis of TOF, which requires early primary repair for long term survival. Being asymptomatic, it is difficult to decide the need for surgery in such cases. After reviewing the literature on management of adult TOF, definitive surgical repair should be the goal. In such scenario, if another cardiac lesion is also present, then it becomes essential to address all the cardiac lesions during a single surgery.

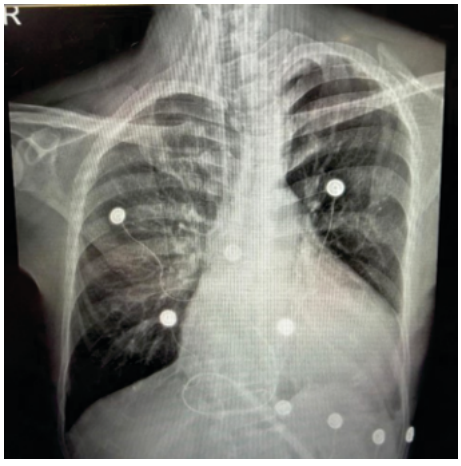

Figure 1: Chest X ray PA View.

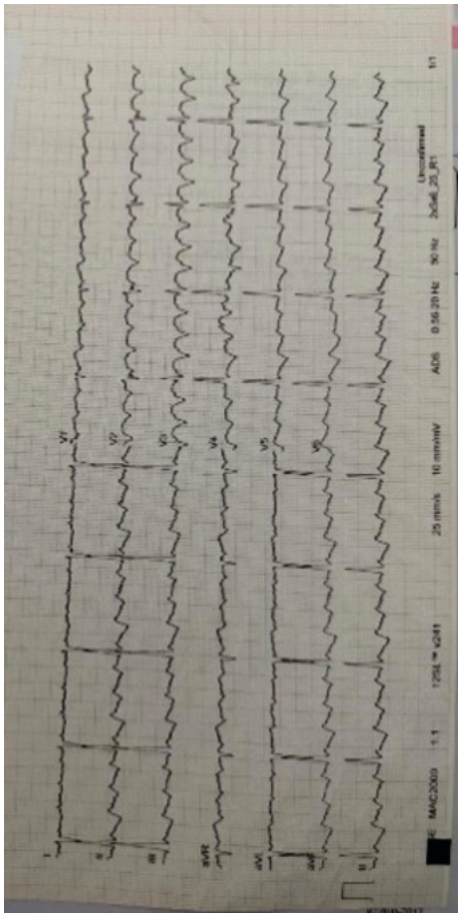

Figure 2:12 Lead Standardized

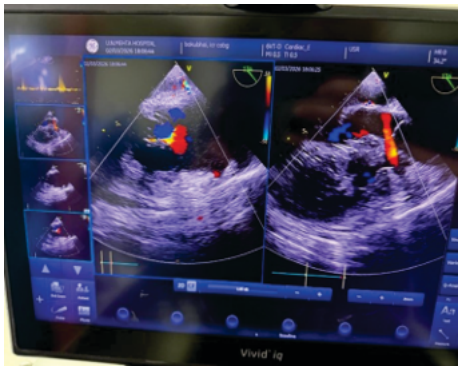

Figure 3: Transesophageal Echo Showing Sub-aortic VSD With Infundibular Stenosis.

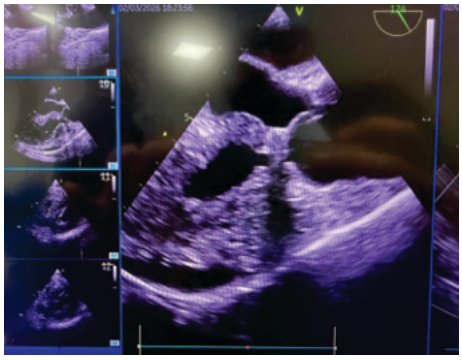

Figure 4: Post Operative Transesophageal Echo Showing VSD Closure with Patch and Wide Open RVOT.

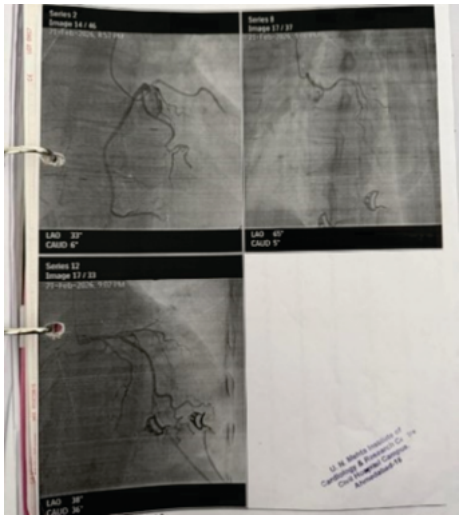

Figure 5: Images of CAG Showing Coronary Lesions.

REFERENCES

1. Warfaa, A. A., Said, A. I., Abdulahi, M., & Hassan, M. S. (2025). Surviving Unrepaired Tetralogy of Fallot to 43 Years in a Low-Resource Setting: The Oldest Reported Case from Somalia. International Medical Case Reports Journal, 18, 827–835. https://doi.org/10.2147/IMCRJ.S529873
2. Van Arsdell GS, Maharaj GS, Tom J, et al. What is the optimal age for repair of tetralogy of Fallot? Circulation. 2000;102(19). doi:10.1161/circ.102.suppl_3.iii-123
3. Bernier PL, Stefanescu A, Samoukovic G, Tchervenkov CI. The challenge of congenital heart disease worldwide: epidemiologic and demographic facts. Semin Thorac Cardiovasc Surg Pediatr Card Surg Ann. 2010;13(1):26–34. doi:10.1053/j.pcsu.2010.02.005
4. Campbell M. Natural history of cyanotic malformations and comparison of all common cardiac malformations. Br Heart J. 1972;34(1):3–8. doi:10.1136/hrt.34.1.3
5. Roman S, Castellarin M. A uniquely compensated boot-shaped heart: a case of unrepaired tetralogy of Fallot with delayed symptom onset in adulthood. Chest. 2020;158(4):A280. doi:10.1016/j.chest.2020.08.281
6. Bailliard F, Anderson RH. Tetralogy of Fallot. Orphanet J Rare Dis. 2009;4(1). doi:10.1186/1750-1172-4-2
7. Haranal MY, Xavier J, Jawali V, Madkaiker A. Tetralogy of Fallot with acquired coronary artery disease—An unusual presentation, J Card Surg, 2016, 725–729, DOI: 10.1111/jocs.12851
8. Rammohan M, Balram Airan, Anil Bhan et al. Total Correction of tetralogy of fallot in adults – surgical experience. International Journal of Cardiology. 1998; 63(2): 121-128. doi: 10.1016/S0167-5273(97)00279-9
9. Abraham KA, Cherian G, Rao VD, Sukumar IP, Krishnaswami S, John S. Tetralogy of Fallot in adults. A report on 147 patients. Am J Med. 1979 May;66(5):811-6. doi: 10.1016/0002-9343(79)91121-5. PMID: 155988.
10. Kirklin JW, Blackstone EH, Jonas RA, Shimazaki Y, Kirklin JK, Mayer JE Jr, Pacifico AD, Castaneda AR. Morphologic and surgical determinants of outcome events after repair of tetralogy of Fallot and pulmonary stenosis. A two-institution study. J Thorac Cardiovasc Surg. 1992 Apr;103(4):706-23. PMID: 1548913.
11. Gregory Y.H. Lip, Shyam P. Singh. Arrhythmias in adults following repair of tetralogy of fallot. The American Journal of Cardiology. 2001; 88(8): 936. DOI: 10.1016/S0002-9149(01)01868-9
12. Egbe, A, Connolly, H. CORONARY ARTERY DISEASE IN ADULTS WITH TETRALOGY OF FALLOT. JACC. 2019 Mar, 73 (9_Supplement_1) 653. https://doi.org/10.1016/S0735-1097(19)31261-6
13. Phillips S, Pirics M. Congenital heart disease and reproductive risk: an overview for obstetricians, cardiologists, and primary care providers. Methodist DeBakey cardiovascular journal. 2017 Oct;13(4):238.
14. Kaur KS, Gupta ML, Rajput HS, Sajan C. Tetralogy of Fallot in adult–uncorrected and rare presentation: a case report. J Young Pharm. 2023 Jan;15(1):189-92.
15. Schäfer M, Mawad W. Advanced imaging technologies for assessing tetralogy of Fallot: insights into flow dynamics. CJC Pediatr Congenit Hear Dis. 2023;2(6):380–392. doi:10.1016/ j.cjcpc.2023.09.011
16. Bhattarai P, Karki M, Purewal JK, Devarakonda Kumar. Unrepaired tetralogy of Fallot: a tale of delayed presentation and limited access to care. Chest. 2023;164(4): A386–A387. doi:10.1016/j.chest.2023.07.316