

Surviving Unrepaired Tetralogy of Fallot to 43 Years in a Low-Resource Setting: The Oldest Reported Case from Somalia

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Introduction: Tetralogy of Fallot (ToF) is a congenital heart defect characterized by a combination of four anomalies: right ventricular hypertrophy, ventricular septal defect (VSD), right ventricular outflow tract (RVOT) obstruction, and an overriding aorta. While surgical interventions have significantly improved the prognosis of ToF, cases of undiagnosed or untreated ToF in adulthood remain, particularly in low-resource settings. This report highlights the case of an adult patient with uncorrected ToF, underscoring the challenges individuals face in such environments.

Case Report: A 43-year-old male from Somalia presented with recurrent syncope, exertional dyspnea, and a history of congenital heart disease diagnosed in childhood. Due to financial constraints and limited healthcare access, he was unable to undergo surgery. On clinical examination, the patient exhibited cyanosis, grade 3 clubbing, and a pansystolic murmur. Investigations showed secondary polycythemia, right ventricular hypertrophy on ECG, and a large VSD with an overriding aorta. Echocardiography confirmed these findings, revealing the anatomical abnormalities typical of ToF.

Discussion: Untreated ToF often leads to significant morbidity, including worsening cyanosis, syncope, and heart failure. In this case, delayed surgical intervention and lack of resources contributed to the progression of symptoms. The patient's case emphasizes the importance of early diagnosis and surgical repair, particularly in resource-limited settings where access to advanced care is limited.

Conclusion: This case highlights the critical need for early intervention and improved healthcare access for individuals with congenital heart defects in low-income settings. Timely diagnosis, surgical repair, and ongoing care can substantially improve outcomes for patients with ToF.

Keywords: Tetralogy of Fallot, congenital heart disease, ventricular septal defect, right ventricular outflow tract, adult diagnosis

Introduction

Tetralogy of Fallot (ToF) is a congenital cyanotic heart defect, first described in detail by Étienne-Louis Fallot in 1888. It is characterized by a combination of four distinct anatomical abnormalities: right ventricular (RV) hypertrophy, a ventricular septal defect (VSD), obstruction of the right ventricular outflow tract (RVOT), and an overriding aorta. These structural anomalies result in altered hemodynamics, reducing pulmonary blood flow and systemic cyanosis.¹ Early primary repair of Tetralogy of Fallot (ToF) has been advocated since the 1970s and is now routinely performed with excellent outcomes.² This approach has become the standard of care, as it promptly addresses the anatomical abnormalities and associated hemodynamic consequences, reducing long-term morbidity and mortality. However, despite the widespread adoption of early surgical intervention, occasional cases of patients who have not undergone repair survive into adulthood.^{3,4} However, delaying the diagnosis and late intervention are highly associated with poor outcomes.⁵ According to large observational series, 24% of individuals with an uncorrected TOF die before the age of 10, and only 4% survive beyond their thirties.^{6,7} We present a unique case of an individual with uncorrected Tetralogy of Fallot (TOF) who has survived into adulthood with a relatively preserved quality of life despite the absence of surgical

intervention. The lack of corrective surgery was primarily due to financial constraints and limited access to specialized cardiovascular care. The patient resides in Somalia, a low-income country where healthcare infrastructure remains underdeveloped, and access to advanced medical and surgical interventions is severely restricted. This case underscores the significant challenges faced by individuals with congenital heart disease (CHD) in resource-limited settings. It highlights the urgent need for improved access to pediatric and adult congenital cardiac care in such environments.

Case Presentation

A 43-year-old male presented with a history of recurrent syncope, reporting three episodes over the past six months. The most recent episode occurred at approximately 3:00 AM, during which the patient was found lying unresponsive on his bed by his family and was subsequently rushed to the hospital. The patient has a longstanding history of exertional dyspnea dating back to childhood, which was particularly pronounced during activities such as climbing hills. Due to his family's nomadic lifestyle, he was often transported on camels during travels to accommodate his physical limitations. Additionally, the patient reported episodes of epistaxis and hemoptysis, further complicating his clinical picture.

The patient's past medical history includes a diagnosis of congenital heart disease of unknown type in 1997. Despite being referred to a cardiac center in Djibouti, a neighboring country, for further management, financial constraints prevented him from accessing specialized care. Over the years, the patient has experienced progressive exertional dyspnea, which has significantly impacted his quality of life and occupational capacity. Initially, he worked in charcoal production and later as a painter, but he was forced to retire due to worsening symptoms. He is the father of six children and has been the primary provider for his family.

Physical Examination

General Appearance

The patient appeared to be in no acute distress and exhibited a normal overall appearance without signs of cachexia.

Vital Signs Were

Blood pressure 110/60 mmHg, heart rate 80 beats/minute, respiratory rate 18 breaths/minute, temperature 35.8 °C, and oxygen saturation 85% on room air.

General Examination

- **Clubbing:** Grade 3 clubbing was observed see [Figure 5](#).
- **Cyanosis:** No cyanosis was noted.
- **Edema:** No peripheral edema was present.

Cardiovascular System (CVS)

- S1 and s2 are audible with a **pansystolic murmur**, graded 3/6 in intensity, was auscultated. The murmur was most prominent in the **tricuspid and mitral areas** but was not associated with a palpable thrill.

Abdomen

- The abdomen was soft and non-tender on palpation.
- No abdominal swelling or organomegaly was 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.

Investigations

Complete Blood Count (CBC)

- **Polycythemia:** Hemoglobin (Hb) level was elevated at **21 mg/dL**, indicative of secondary polycythemia, likely due to chronic hypoxemia associated with congenital heart disease.
- **Thrombocytopenia:** A Low platelet count was noted, possibly related to chronic disease or other underlying factors.

Echocardiographic evaluation of the patient revealed a large malalignment subaortic ventricular septal defect (VSD) with an overriding aorta of less than 50%, indicative of abnormal conal septal development. Right ventricular hypertrophy (RVH) was observed, consistent with chronic pressure overload. Infundibular pulmonary stenosis was noted, contributing to right ventricular outflow obstruction. Additionally, an interatrial septal (IAS) aneurysm was identified, suggesting a structural abnormality that may have clinical significance (Figures 1 and 2).

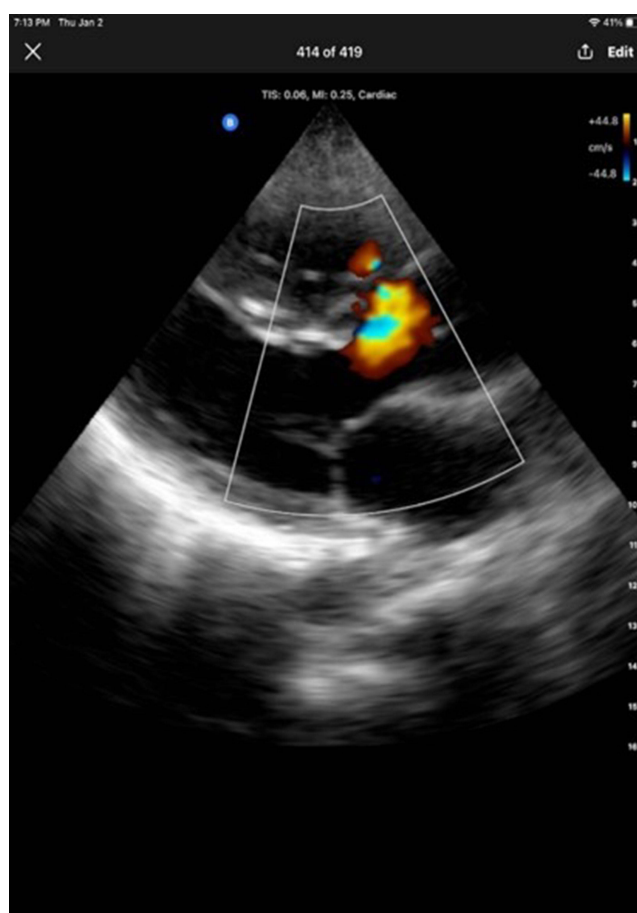


Figure 1 Is a long-axis view demonstrating the VSD with color Doppler imaging, showing flow across the defect penetrating the interventricular septum.

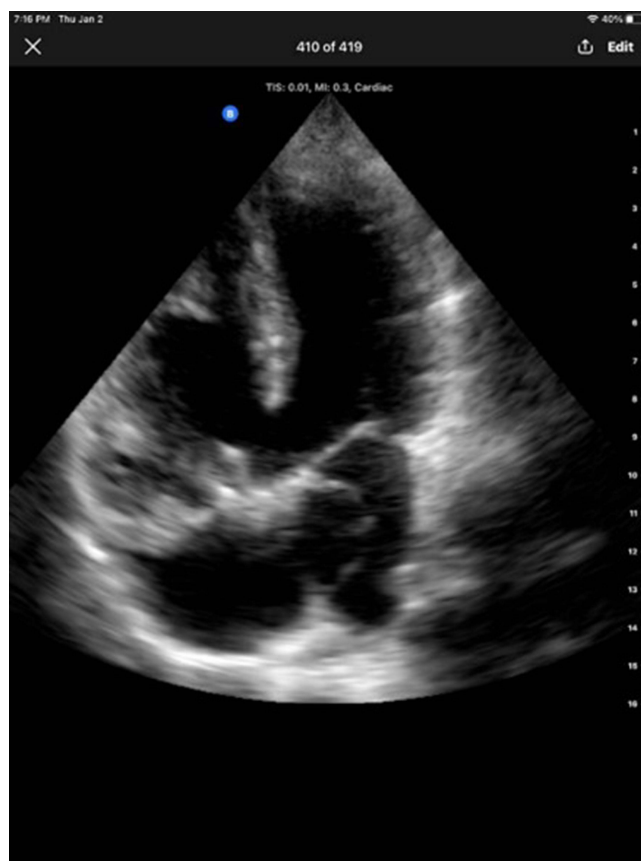


Figure 2 Is an apical four-chamber view providing additional visualization of the VSD.

Assessment

- **Tetralogy of Fallot (ToF):** Known congenital heart defect with concerns for arrhythmias due to recent syncope.
- **Possible Old Inferior Myocardial Infarction (MI):** ECG shows pathological Q waves in inferior leads, possibly due to chronic hypoxemia or coronary artery disease.
- **Secondary Polycythemia:** Elevated hemoglobin (21 mg/dL) as a compensatory response to chronic hypoxemia, increasing thromboembolic risk.
- **Thrombocytopenia:** Low platelet count, possibly secondary to chronic disease or polycythemia.

Plan

- **Medical Management:**
 - Aspirin (81 mg daily): To reduce thromboembolic risk from severe polycythemia and possible atherosclerosis indicated by ECG.
 - Atorvastatin (20 mg daily): Secondary prevention for suspected old inferior MI despite unknown lipid levels.
 - Hydroxyurea (500 mg daily): To control symptomatic secondary polycythemia by lowering erythropoiesis and blood viscosity.
 - Home Oxygen Therapy (as needed): To relieve chronic hypoxemia symptoms, especially exertional dyspnea.
- **Phlebotomy:** Weekly 250 mL blood transfusion to manage polycythemia, with caution due to thrombocytopenia.
- **Further Investigation:** Holter monitoring for arrhythmias.
- **Referral for Cardiac Surgery:** Consideration for pulmonary valve replacement or complete repair.
- **Lifestyle & Follow-up:** Avoid strenuous activity, and regularly monitor hemoglobin, platelets, and cardiac function.

Electrocardiogram (ECG)

- **Rate:** 75 beats per minute.
- **Rhythm:** Sinus rhythm.
- **Axis:** Extreme right axis deviation.
- **P Wave:** P-pulmonale (tall, peaked P waves in leads II, III, and aVF) suggests right atrial enlargement.
- **Q Wave:** Pathological Q waves were observed in the **inferior leads (II, III, and aVF)**, suggesting a possible old inferior wall myocardial infarction.
- **R Wave:**
 - a. Prominent R wave in **V1** and **aVR**.
 - b. Deep S wave in **V5** and **V6**.
 - c. Poor R-wave progression across the precordial leads ([Figure 3](#)).
- **Findings:**
 - **Right Ventricular Hypertrophy (RVH):** Suggested by the prominent R wave in V1 and deep S wave in V5/V6.
 - **P-pulmonale:** Indicative of right atrial enlargement, likely secondary to pulmonary hypertension or chronic right heart strain.
 - **Old Infarction:** Pathological Q waves in the inferior leads raise the possibility of a prior inferior wall myocardial infarction.

Chest X-Ray

- The chest X-ray reveals characteristics of Tetralogy of Fallot, notably a boot-shaped heart caused by enlargement of the right ventricle ([Figure 4](#)).

Physical Appearance

- Hand of the patient showing evidence of digital clubbing ([Figure 5](#)).

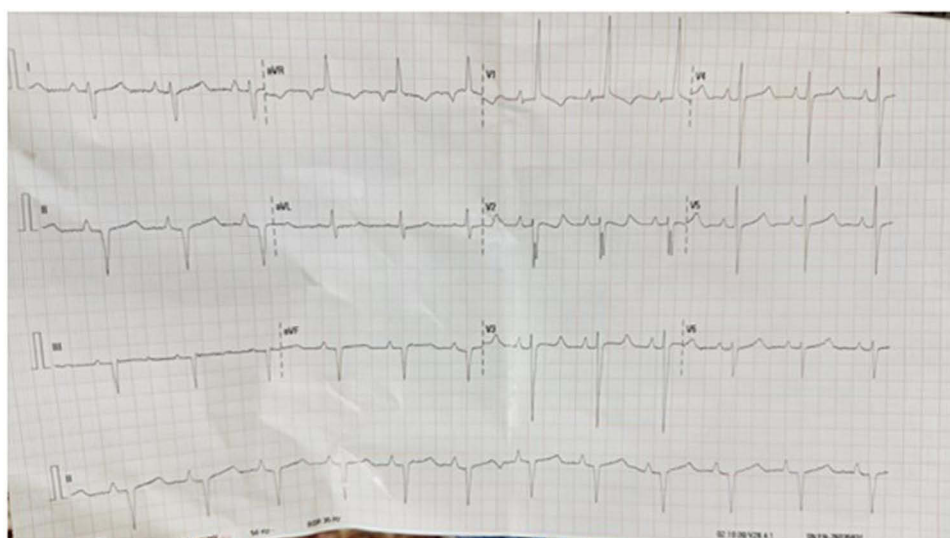


Figure 3 ECG showing right ventricular hypertrophy, right atrial enlargement (P-pulmonale), and pathological Q waves in inferior leads suggest an old inferior wall myocardial infarction.



Figure 4 Chest X-ray showing features of tetralogy of Fallot, including a boot-shaped heart due to right ventricular hypertrophy.



Figure 5 Hand of the patient showing evidence of digital clubbing.

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. The recurrence risk in affected families is approximately 3%.⁸

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.⁹

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. Emerging techniques such as 4D-flow MRI enhance diagnostic precision by enabling dynamic flow analysis, offering insights into adverse hemodynamic patterns, and assisting with risk stratification and intervention planning.¹⁰ While cardiac catheterization remains the gold standard for direct hemodynamic measurements and detailed visualization of pulmonary arteries or complex vascular anomalies like Major Aortopulmonary Collateral Arteries (MAPCAs), its use is often limited in low-resource environments. Therefore, ECG and echocardiography remain indispensable tools for initial evaluation and follow-up in such settings.⁹ In our patient, ECG and echocardiography were indispensable for diagnosing ToF.

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.¹¹

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 the electrocardiogram (ECG) of our patient, the presence of QS waves in the inferior leads raised suspicion of an inferior myocardial infarction (MI). The relationship between myocardial infarction and Tetralogy of Fallot (TOF) is reported in the literature. A case from Turkey described a patient with TOF who experienced an MI, providing valuable insights into this uncommon association. In that case, the absence of resting cyanosis suggested a minor right-to-left shunt, potentially explaining the patient's prolonged survival. However, following the myocardial infarction, both ventricles rapidly deteriorated, leading to severe heart failure and early mortality.¹²

The patient, a 43-year-old man, was admitted with chest pain, dyspnea, and diaphoresis. His medical history revealed dyspnea, exertional cyanosis, and palpitations since childhood. He had been diagnosed with TOF 13 years earlier, at which time cardiac catheterization was performed. However, he declined TOF corrective surgery, which may have contributed to his later cardiovascular complications.¹² This case underscores the complex interplay between congenital heart defects and ischemic heart disease, highlighting the need for careful cardiovascular risk assessment in patients with uncorrected TOF. In the literature, several cases also describe similar situations, where the coexistence of TOF and MI significantly worsens the prognosis.¹³

In our patient, syncope raised suspicion of an underlying arrhythmia. Syncope is a concerning symptom in individuals with repaired Tetralogy of Fallot (TOF). It is often associated with arrhythmias and conduction abnormalities, common long-term sequelae of surgical repair. While advancements in surgical techniques have significantly improved survival rates, residual cardiac abnormalities, scarring from patch material, atriotomy, and ventriculotomy contribute to the development of rhythm disturbances.¹⁴ However, cases of arrhythmias have also been reported in patients with unrepaired TOF.¹⁵ Similar pathophysiological mechanisms could potentially account for the syncopal episode observed in our patient.

Ideally, ambulatory ECG monitoring—such as a Holter monitor—would have been indicated to assess for transient arrhythmias that might not be captured on routine ECG. Contemporary diagnostic strategies often rely on a spectrum of prolonged monitoring modalities, including traditional 24–48-hour Holter monitors, external loop recorders (ELRs), wearable patch devices, and implantable cardiac monitors (ICMs) for prolonged surveillance in select patients. These tools are critical for establishing symptom–rhythm correlation, especially when symptoms are infrequent or unpredictable.¹⁶ As Carrington et al (2022) emphasize, the choice of monitoring modality is primarily dictated by the frequency and nature of symptoms, with longer-duration or patient-activated monitors being particularly advantageous for episodic events. Unfortunately, such diagnostic tools were unavailable due to limitations inherent in our resource-constrained setting. Consequently, only a standard one-minute 12-lead ECG was performed, which showed no evidence of acute ischemia or arrhythmic disturbances at the recording time.

Conclusion

Tetralogy of Fallot (TOF) is a congenital heart defect with significant long-term risks, particularly in uncorrected cases. Our patient's ECG suggested an inferior myocardial infarction, while syncope raised suspicion of arrhythmias, though Holter monitoring was unavailable. This case underscores the critical need for early diagnosis, timely surgical intervention, and continuous cardiac surveillance. In resource-limited settings, improving access to echocardiography and ambulatory ECG monitoring is essential for mitigating complications and improving outcomes in patients with uncorrected TOF.

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Manuscript Submission

We confirm that this manuscript is original and has not been submitted elsewhere for publication.

Ethical Approval

The Ethical Committee of Amoud University granted ethical approval for this study, including permission for publication (Reference: 0100-AU-REC-2025).

Consent for Publication

The patient provided written informed consent after receiving a detailed explanation of the study's purpose, procedures, and potential for publication. This consent included permission to publish all clinical details and associated images in this report. Patient anonymity has been preserved.

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Author Contributions

All authors contributed significantly to developing this case report, including the conception and interpretation of clinical findings. They participated in drafting, revising, or critically reviewing the manuscript; approved the final version to be published; agreed on the journal to which the case report was submitted; and took full responsibility for all aspects of the work.

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Disclosure

The authors declare that there are no conflicts of interest regarding the content or publication of this manuscript.

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