

# Uncorrected Tetralogy of Fallot with Pulmonary Atresia in an Asymptomatic 35 Year Old Nigerian Man

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## Abstract

Tetralogy of Fallot is the commonest cyanotic congenital heart disease after the neonatal period, accounting for 10 - 26.2% of congenital heart diseases in Nigeria. Structural abnormalities found in tetralogy of fallot could explain associated adverse haemodynamic changes. It is imperative to document a case of an uncorrected tetralogy of Fallot with pulmonary atresia in a 35-year old Nigerian African man, considering the associated poor/high morbidity and mortality. Explanations for his longevity are left ventricular hypertrophy and systemic-pulmonary collateral circulation.

**Keywords:** *Tetralogy of Fallot, Pulmonary Atresia, Nigerian/African, Uncorrected, Asymptomatic*

## Introduction

Tetralogy of Fallot (ToF) is the commonest form of cyanotic congenital heart disease accounting for about 10% -26.2% of all cases of congenital heart diseases in Nigeria.<sup>[1]</sup> It consists of interventricular septal defect, right ventricular outflow tract obstruction, an overriding aorta and right ventricular hypertrophy. Without corrective surgery, few patients with ToF reach adulthood with an average life expectancy of 12 years.<sup>[2,3]</sup> Prognosis of patients with ToF with pulmonary atresia (PA) is worse than that in ToF with pulmonary stenosis as less than 10% of patients with ToF and PA are expected to be alive at age 20 years.<sup>[4-6]</sup>

The degree of symptoms depends on the degree of RVOT obstruction and additional source of blood supply to the pulmonary circulation.<sup>[7]</sup> We present the case report of a 35-year old man with an uncorrected TOF with pulmonary atresia having obtained consent to publish from him. We also review the possible factors contributing to his longevity. There is no conflict of interest in this publication.

## Case Report

He is a 35- year- old civil servant who was referred to us by the haematologist on account of polycythaemia (PCV-76%) and features suggestive of cyanotic congenital heart disease. The patient had no history of dyspnea with any degree of exertion, also able to climb stairs or hills without any difficulty; no orthopnea, or paroxysmal nocturnal dyspnea. There was no history of cough, pleurisy or chest pain, palpitation, presyncope or syncope. No history of headache, blurring of vision or tinnitus.

There was a history of dyspnea on exertion and effort intolerance during his childhood. He admitted to frequent squatting to relieve

episodes of breathlessness following moderate exertion often noticed while playing football and this started at 4 years of age and stopped spontaneously at about 11 years without any intervention. Since then, he has been asymptomatic. He is not a known patient with hypertension, diabetes mellitus or bronchial asthma. No family history of heart disease. There is history of chronic apical periodontitis two years ago which culminated in the removal of two teeth. He is married to one wife with two children.

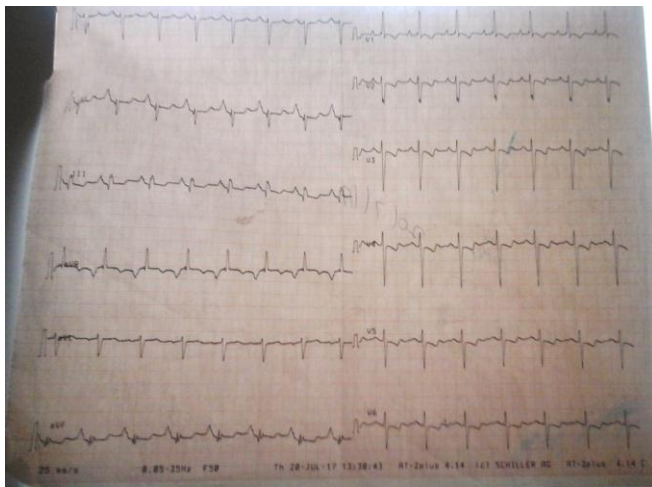
General examination revealed an underweight (BMI-16.5kg/m<sup>2</sup>), plethoric patient who had both central and peripheral cyanosis with grade 4 digital clubbing. There was no peripheral oedema. He had poor oral hygiene. Oxygen saturation was 81%.

Cardiovascular system examination revealed a pulse of 70 per minute, regular and of normal volume. The blood pressure was 120/94 mmHg and jugular venous pressure was normal. The apex beat was located at the 5th left intercostal space in the mid clavicular line and there was left parasternal heave but no thrills. On auscultation, there were first and second heart sounds at the apex, an additional fourth heart sound at the 4th left parasternal area, and grade 3/6 pansystolic murmur which was at the left lower sternal edge. He had an accentuated A2 sound. He had pectus carinatum with evidence of hyperinflated lungs. The chest was clinically clear. The neurological, abdominal and musculoskeletal systems were normal.



Chest radiograph (figure 1) showed: an enlarged cardiac silhouette with right ventricular preponderance, right atrial enlargement, left-sided dilated aortic arch, small main pulmonary artery, and widespread dilated pulmonary vessels with upper lobe diversion.

Electrocardiography (figure 2) showed sinus rhythm, extreme QRS axis deviation, right atrial abnormality, biventricular hypertrophy with secondary ST-T wave abnormalities in anterior and inferior leads



**Figure 2 showing electrocardiograph of the patient with sinus rhythm, right atrial abnormality and biventricular hypertrophy. (speed -25mm/sec, amplitude-5mm/mV)**

Echocardiography (figure 3) showed: a membranous ventricular septal defect measuring 1.9cm with a bi-directional colour flow and an overriding dilated aorta (with aortic diameter of 3.9cm), interventricular septal wall hypertrophy of 1.6cm in diastole, and normal left atrial (2.2cm) and left ventricular cavity (4.4cm) with a slightly depressed left ventricular ejection fraction (52%). There was also right ventricular hypertrophy (RVH) with the right ventricular free wall of 12cm, and a normal longitudinal function with a tricuspid annular systolic excursion (TAPSE-1.36cm); dilated right atrium (RA) with an area of 25.2 cm<sup>2</sup> and dilated inferior venal cava (IVC-2.96cm) with less than 50% collapse during inspiration. The pulmonary valve was thickened with a small pulmonary artery. The pulse wave was reversed in both right and left ventricular inflow.



**Figure 3 showing echocardiogram of an adult patient with Tetralogy of Fallot**

The serum electrolytes, urea and creatinine were within normal limits and complete urinalysis was normal.

Supportive and preventive management were instituted for the patient. This included counseling and education on nature of the disease, infective endocarditis prophylaxis advised, oral care, and successive phlebotomy. He had referral for cardiothoracic consultation.

## Discussion

Several reasons have been put forward to explain increased longevity in patients with ToF.<sup>[8-10]</sup> One of these is gradual development of RVOT obstruction.<sup>[9]</sup> This patient experienced episodes of severe hypoxia only during severe physical activities which appeared to have happened over a period of about 7 years during his childhood. This could be as a result of hypoplastic pulmonary trunk with gradual development of RVOT obstruction over 7 years alongside gradual opening up of aortopulmonary circulation. Therefore, during adulthood he remained asymptomatic at higher levels of activities with full development of aortopulmonary circulation and had been fully adapted to chronic hypoxia. Distance covered during 6-minute walk test was 450metres without development of dyspnea and arterial oxygen saturation at room air was 81%.

The presence of aortopulmonary circulation was evidenced on chest X-ray with prominent bronchovascular markings in the right lung and left upper lung zones. There was pulmonary oligoemia in the left mid and lower lung zones. Various patterns of aortopulmonary circulation have been described by Santos MA *et al*<sup>[11]</sup> in an extensive work on angiographic patterns of pulmonary circulation in ToF with pulmonary atresia.<sup>[11]</sup> Extensive arborization of aortopulmonary collateral circulation emanating from the hilar could probably explain his asymptomatic state.

Presence of RVH suggests pressure overload from RVOT obstruction while the dilatation of right ventricle and atrium points to volume overload which arose from shunting through the interventricular septal defect. Colour doppler showed bi-directional blood flow across ventricular septal defect (VSD). This is consistent with pattern of pressure gradient across the defect during different phases of cardiac cycle. Right to left shunting is due to pressure overload due to RVOT obstruction while left to right shunting is the effect of left ventricular hypertrophy (LVH)

development. LVH is an adaptive response that often develops late during the course of the disease condition, as a result of volume overload. Subsequently, LVH reduces the extent of right to left shunt.<sup>[8]</sup>

Improved technology and knowledge in surgery, particularly in congenital heart disease, has improved life expectancy of patients with congenital heart diseases. This can be said of developed countries, unlike developing nations like Nigeria. All patients with ToF will benefit from surgery which eventually may be more than once during their lifetime. Expertise is available in the country, though very limited, likewise investigation modalities such as cardiac magnetic resonance imaging, cardiac catheterization and angiography.

To the best knowledge of the authors, this is the first documented case of uncorrected asymptomatic ToF with pulmonary atresia in an adult Nigerian. Previous reports in Nigeria<sup>[2,12]</sup> and possibly in Africa, were on survivors of uncorrected TOF with pulmonary stenosis in adult.

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