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Case Report

Uncorrected Tetralogy of Fallot with severe pulmonary stenosis in an adult: a case report

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ABSTRACT

Tetralogy of Fallot (TOF) is a complicated cyanotic congenital heart problem that is uncommon in adults where it is uncorrected. The present case report tells about a patient of 35 years, who had uncorrected TOF and severe pulmonary stenosis, who came with complaints of cough and chest pain. Post-contrast CT depicted that main pulmonary artery, right pulmonary artery, and left pulmonary artery were severely constricted (5.3 mm, 5.8 mm, 5.5 mm), ascending aorta showed aneurysmal dilation (4.9 cm) and aorta arch (3.9 cm), and presence of various aortopulmonary collaterals. Other positive results were right ventricular hypertrophy, bilateral perinephric collections, fibrosis, and ground-glass lung opacities, partially attributable to previous tuberculosis. The results indicate the balancing vascular responses that maintain survival, the systemic sequelae of chronic hypoxemia and immediate dangers such as aortic rupture. The case indicates the necessary approach to multidisciplinary management and adds unique knowledge to unresolved TOF adult outcomes.

Keywords: Tetralogy of Fallot, Pulmonary stenosis, Cardiac and vascular, Case report

INTRODUCTION

Tetralogy of Fallot (TOF) is a congenital heart defect characterized by four anatomical abnormalities: a ventricular septal defect (VSD), right ventricular outflow tract obstruction (typically pulmonary stenosis), right ventricular hypertrophy, and an overriding aorta.[1] With an incidence of approximately 0.34 per 1000 live births, TOF is one of the most common cyanotic congenital heart diseases.[2] The severity of pulmonary stenosis varies, ranging from mild to severe, and significantly influences the clinical presentation and management. In severe cases, the obstruction can lead to reduced pulmonary blood flow, cyanosis, and compensatory mechanisms such as the development of aortopulmonary collateral arteries.[3]

Most patients with TOF undergo surgical correction in infancy or early childhood to alleviate symptoms and prevent complications such as progressive cyanosis, heart failure, and arrhythmias.[4] However, a small subset of patients survive into adulthood without surgical intervention, often due to a balanced physiology where the degree of pulmonary stenosis allows sufficient pulmonary blood flow to prevent severe cyanosis.[5]

These patients remain at risk for complications, including right ventricular dysfunction, aortic dilatation, and systemic effects of chronic hypoxemia, such as polycythemia.[6]

This case report presents a 35-year-old male with uncorrected TOF and severe pulmonary stenosis who presented with cough and chest pain. Patient's history of tuberculosis adds complexity to his clinical management, highlighting challenges of managing multisystem complications in adults with congenital heart disease.

CASE REPORT

A 35-year-old male with a known history of TOF and a past medical history of tuberculosis diagnosed six years prior presented to the hospital with complaints of cough and chest pain. The patient had no history of surgical intervention for his congenital heart defect. The clinical presentation prompted further diagnostic evaluation.

A post-contrast computed tomography (CT) scan of the chest and abdomen revealed the following findings:

Thoracic findings

Cardiac and vascular

Severe stenosis of main, right, and left pulmonary arteries, with diameters of 5.3, 5.8, and 5.5 mm, respectively, indicating significant obstruction to pulmonary blood flow and suggestive of severe pulmonary hypertension.

Aneurysmal dilatation of the ascending aorta (4.9 cm) and aortic arch (3.9 cm), significantly above normal diameters, with the descending thoracic aorta measuring 2.3 cm.

Multiple aortopulmonary collaterals around the carinal region, aortopulmonary window, left prevascular region, and precarinal and subcarinal regions, providing alternative pathways for pulmonary blood flow.

Right ventricular hypertrophy, consistent with chronic pressure overload due to severe pulmonary stenosis.

An enhancing lesion in the subpleural region of the anterior segment of the right upper lobe, possibly representing collateral vessels, adjacent to thrombosis in the right subclavian vein.

Pulmonary

Low attenuation regions in multiple lung segments (right upper lobe, right middle lobe, right lower lobe, left upper lobe, left lingular segment, and left lower lobe), suggestive of air trapping or perfusion defects, requiring correlation with expiratory or perfusion scans.

Regions of fibrosis in the anterior segment of the right lower lobe, posterior segment of the left lower lobe, and foci in the right middle lobe, potentially related to prior tuberculosis.

Subpleural ground-glass opacity in the posterior segment of the left lower lobe.

Minimal bilateral pleural effusion.

Abdominal findings

Bilateral perinephric collections, measuring 2.4 cm on the right and 2.1 cm on the left, possibly related to polycythemia or pulmonary arterial hypertension.

Faint nephrographic defects in both kidneys during the arterial phase, necessitating further evaluation with a complete CT kidneys, ureters, and bladder (KUB) with contrast.

Perinephric fat stranding with thickening of the anterior, posterior, and lateroconal fascia.

Fluid in the right anterior pararenal space and minimal fluid in the left anterior pararenal space.

No specific treatment or outcome data were available at the time of evaluation.

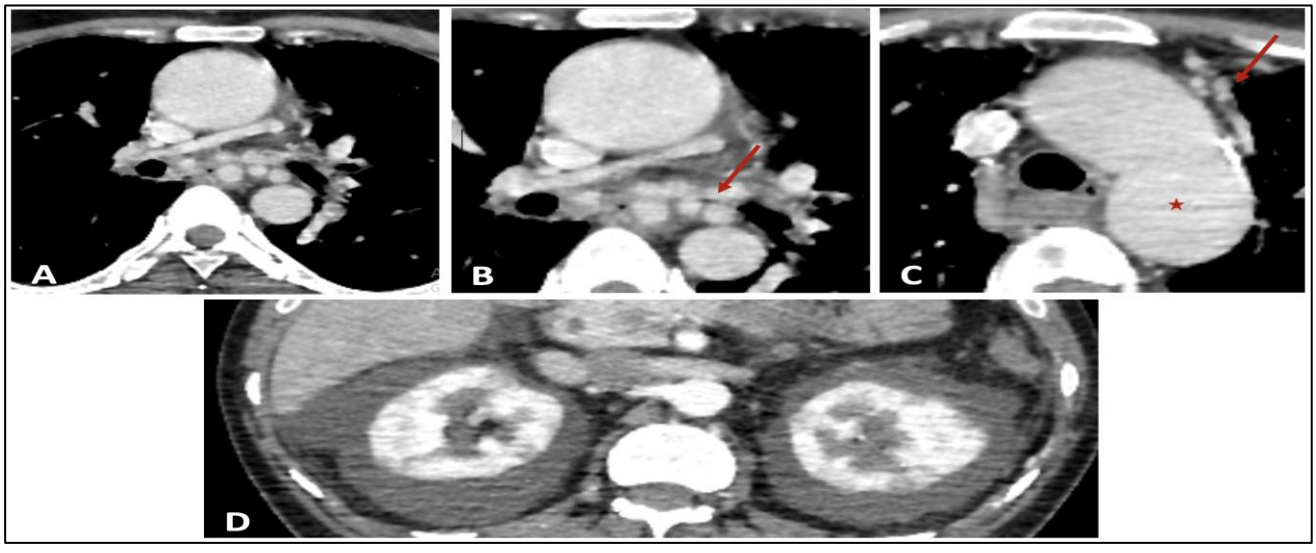

Figure 1 (A-D): Post-contrast CT axial images showing: A-severely stenotic main pulmonary artery and right pulmonary artery. B-multiple aorto-pulmonary collaterals around the carinal region (red arrow). C-aneurysmally dilated aortic arch (red star) and left prevascular region collaterals (red arrow). D-bilateral perinephric collection.

DISCUSSION

The survival of a patient with uncorrected TOF into adulthood is rare, particularly with severe pulmonary stenosis. The degree of pulmonary stenosis in TOF determines the extent of right-to-left shunting through the VSD and the severity of cyanosis. In this patient, the severe stenosis of the main, right, and left pulmonary arteries, with diameters of 5.3 mm, 5.8 mm, and 5.5 mm, respectively, suggests significant obstruction to pulmonary blood flow. The presence of multiple aortopulmonary collaterals indicates a compensatory mechanism to maintain pulmonary perfusion, which likely contributed to the patient's survival into adulthood without surgical correction.[7] Historical data suggest that only a small percentage of patients with uncorrected TOF survive beyond childhood, with life expectancy significantly reduced without intervention.

The patient's presenting symptoms of cough and chest pain may be attributed to several factors. The severe pulmonary stenosis and resultant pulmonary hypertension could lead to right ventricular strain, contributing to chest pain. Additionally, the lung parenchymal abnormalities, including fibrosis and ground-glass opacities, may reflect sequelae of the patient's previous tuberculosis infection or chronic hypoxemia secondary to TOF.[8] The minimal bilateral pleural effusion could be related to heart failure or an inflammatory process, further complicating the clinical picture.

A critical finding in this case is the aneurysmal dilatation of the ascending aorta (4.9 cm) and aortic arch (3.9 cm). Aortic root dilatation is a well-documented complication in TOF, observed in both repaired and unrepaired cases, and is attributed to intrinsic aortopathy and increased hemodynamic stress from the overriding aorta.[9] The risk of aortic dissection or rupture increases with diameters approaching 5.5 cm, though in patients with congenital heart disease, earlier intervention may be considered due to the heightened risk.[10] The aortic dilatation in this patient likely contributes to the chest pain and warrants urgent evaluation by a cardiothoracic surgeon to assess the need for surgical intervention, such as aortic root replacement or repair.[11]

The bilateral perinephric collections are another significant finding. The collections could be hematomas resulting from secondary polycythemia, a common consequence of chronic hypoxia in cyanotic heart disease, which increases the risk of spontaneous bleeding due to hyperviscosity.[12]

The lung parenchymal findings, including fibrosis, ground-glass opacities, and low attenuation regions, suggest a complex interplay of infectious and cardiac-related pathologies. Chronic hypoxia and pulmonary hypertension associated with TOF can lead to pulmonary vascular disease and infarction, contributing to parenchymal changes. The history of tuberculosis further complicates the differential diagnosis, as residual scarring or reactivation of infection could account for the observed abnormalities. Additional diagnostic workup, such as pulmonary function tests, bronchoscopy, or biopsy, may be necessary to clarify the etiology and guide treatment.

Individualised multidisciplinary approach is needed to manage uncorrected TOF with severe pulmonary stenosis in adulthood. The definitive treatment is surgical correction that entails closure of VSD and the right ventricular outflow obstruction.[13] In long-standing and untreated cases, early surgical intervention is technically unfair because of chronic hypertrophy of the right ventricle, permanent pulmonary hypertension and formation of collaterals. In carefully chosen patients there is the possibility of placing some palliative procedures like the systemic-to-pulmonary artery shunts or balloon pulmonary valvotomy to increase pulmonary blood flow. The aneurysmal dilation of the ascending aorta is aneurysmal (4.9 cm) so there is the need to perform cardiothoracic assessment to have the aortic root replaced or fixed before it ruptures or dissects. There is medical therapy which comprises phlebotomy or hydroxyurea therapy in secondary polycythemia, anticoagulants as thrombosis prophylaxes, diuretics and beta blockers to counter the symptoms of cardiac failure and cardiac arrhythmia. Pulmonary combination of fibrosis and ground-glass opacities will need subsequent monitoring according to CT, pulmonary functions, as well as latent tuberculosis reactivation, as the patient has a history of it. Urinary tract complications that require nephrology review show include bilateral perinephric collections and hydration regimes and observations of hematoma or infection. Long-term cardiology follow-up post-treatment is required and should include monitoring of cardiac performance by echocardiography and CT/MRI imaging of the right ventricular and aortic dimension.[14] During their treatment, patients are supposed to receive endocarditis prophylaxis and vaccination to minimize risks of infections. An ACHD clinic with structured follow up is advised and incorporates the expertise of cardiology, pulmonology, nephrology, and hematology. Deliberate surveillance and prompt interventions are the key issues to enhance survival and quality of life in these rare high-risk adult TOF manifestations.

Managing this patient presents significant challenges due to the multisystem involvement. A multidisciplinary approach involving cardiologists, cardiothoracic surgeons, infectious disease specialists, nephrologists, and pulmonologists is crucial. Potential management strategies include:

Cardiac

Evaluation for surgical correction of TOF, such as relief of pulmonary stenosis and closure of the VSD, though this is complex in adults due to long-standing physiological changes.[15] Palliative procedures to augment pulmonary blood flow may also be considered.[16]

Pulmonary

Further imaging, such as expiratory or perfusion scans, to assess lung abnormalities, and evaluation for active tuberculosis if suspected.

Systemic

Monitoring for complications of polycythemia, such as thrombosis, and managing pulmonary hypertension if present.

The complexity of this case underscores the need for individualized care plans and highlights the importance of long-term follow-up in adults with congenital heart disease. The limited literature on uncorrected TOF in adulthood emphasizes the value of such cases in understanding natural history and optimal management strategies.

CONCLUSION

This case report describes a rare presentation of a 35-year-old male with uncorrected TOF and severe pulmonary stenosis, presenting with cough and chest pain. Imaging revealed severe pulmonary artery stenosis, multiple aortopulmonary collaterals, aneurysmal aortic dilatation, lung parenchymal abnormalities, and bilateral perinephric collections, potentially linked to a history of tuberculosis or polycythemia. These findings highlight the complex interplay of congenital heart disease, infectious complications, and systemic effects. This case contributes to the limited literature on adults with uncorrected TOF and emphasizes the need for comprehensive care and further research into this rare condition.

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