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Case Series

Internal Medicine Section

Clinical Spectrum of Tetralogy of Fallot with Right Ventricular Outflow Tract Variants from Infancy to Adulthood: A Case Series

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ABSTRACT

Tetralogy of Fallot (TOF) with Right Ventricular Outflow Tract (RVOT) variants, such as TOF with pulmonary atresia (10-15% of TOF cases) and TOF with Absent Pulmonary Valve (APV) (3-6% of TOF cases), presents diverse clinical features and management challenges across age groups. The present case series describes a seven-month-old male infant with TOF and APV featuring respiratory distress, a 14-year-old male adolescent with unoperated TOF and pulmonary atresia showing progressive cyanosis, and a 31-year-old female adult with TOF, pulmonary atresia post-bilateral Blalock-Taussig (BT) shunts experiencing exertional dyspnoea. These cases highlight age-specific presentations, diagnostic findings, and tailored management strategies. Early diagnosis, lesion-specific palliation or repair, and lifelong surveillance are essential to reduce long-term complications such as Right Ventricular (RV) dysfunction and arrhythmias, particularly in resource-limited settings.

Keywords: Absent pulmonary valve, Blalock-taussig shunt, Cyanotic congenital heart disease, Pulmonary atresia

INTRODUCTION

The TOF is the most common cyanotic congenital heart disease, characterised by a malaligned ventricular septal defect, RVOT obstruction, overriding aorta, and RV hypertrophy. Variants like TOF with pulmonary atresia and TOF with APV alter pulmonary blood flow and airway dynamics, impacting prognosis [1,2]. Advances in imaging and surgery have enhanced survival to adulthood, yet late presentations persist in resource-limited areas, underscoring the need for the current series to illustrate lifespan management. The present case series describes presentation, features, and management of three cases of TOF variants encountered at a single centre. Written informed consent was obtained from patients/guardians for publication.

CASE SERIES

Case 1

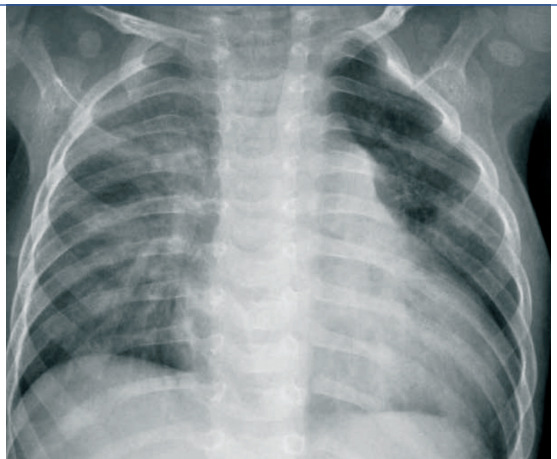
Infant TOF with Absent Pulmonary Valve (APV): A seven-month-old male infant, with normal antenatal scans for mother and an uneventful pregnancy, born out of a non-consanguineous marriage at term by uncomplicated vaginal delivery with birth weight 2.5 kg, was referred to a higher centre soon after birth with heart defects. There was no history of bluish discoloration at birth or thereafter. The mother reported recurrent episodes of rapid, heavy breathing during play, associated with frequent lower respiratory tract infections and wheezing. Feeding was adequate, and there was no developmental delay or failure to thrive.

On general examination, the infant was conscious, afebrile, and acyanotic, with no pallor, icterus, clubbing, pedal oedema, or lymphadenopathy. Oxygen saturation was 93% in the right upper limb. Pulse rate was 120/min, regular and of normal volume, with no radio-radial or radio-femoral delay. Jugular venous pressure was not elevated.

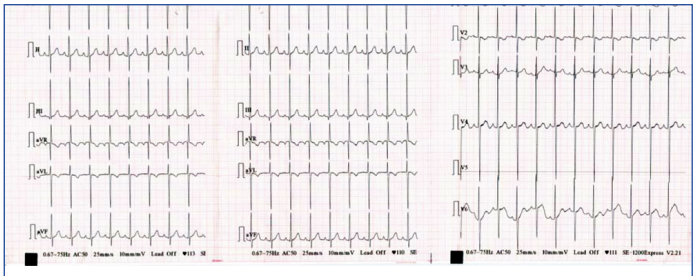
The chest was symmetrical with the trachea in the midline. The apex beat was located in the fifth left intercostal space, about 2 cm lateral to the mid-clavicular line, with associated lateral retraction and precordial bulge. Epigastric impulse was present. There was no parasternal heave, no palpable P2, and no palpable thrill. Percussion revealed normal cardiac borders, with liver dullness in the right fifth intercostal space and dullness in the left second intercostal space.

On auscultation, normal S1 and S2 were heard at the mitral, tricuspid, and aortic areas, without added sounds or murmurs. At the pulmonary area, there was a to-and-fro murmur comprising a low-pitched diastolic component and a high-pitched grade 4 crescendo-decrescendo ejection systolic murmur radiating over the precordium. The second heart sound was single, with P2 not audible. Systemic examination was otherwise unremarkable.

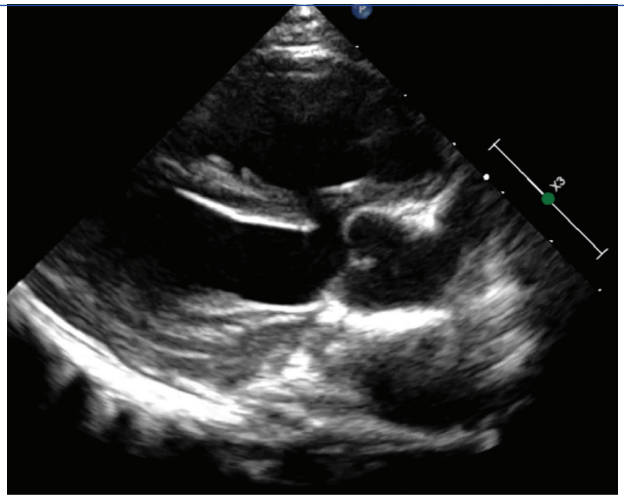
Based on the clinical profile, the diagnosis was congenital heart disease, TOF with APV, in sinus rhythm, Ross class I, with normal left ventricular function and no heart failure. Chest radiograph [Table/Fig-1], electrocardiogram [Table/Fig-2], and echocardiography [Table/Fig-3-5] were consistent with TOF anatomy, APV, and increased pulmonary blood flow. The infant was managed medically with antibiotics, nebulisation, vaccines for prevention of respiratory infections, and surgically with ventricular septal defect closure, along with RVOT reconstruction and pulmonary artery reduction plasty.



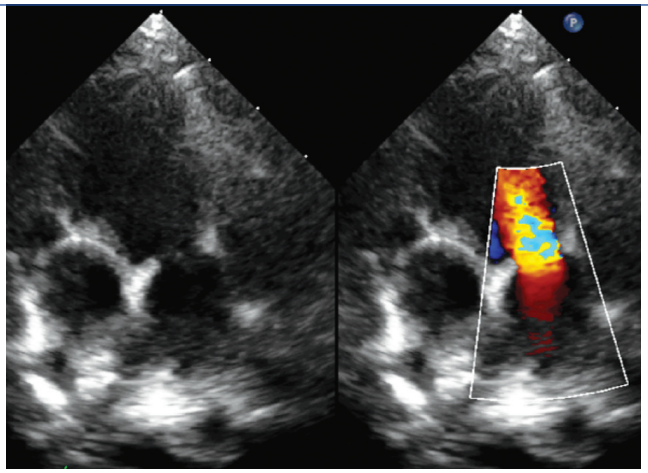
[Table/Fig-1]: X-ray shows massively dilated central pulmonary arteries (appearing "aneurysmal") and often hyperinflated lungs with bronchial compression, leading to atelectasis or lobar emphysema, along with cardiomegaly from the dilated RV, distinct from the typical "boot-shaped" heart of standard TOF.



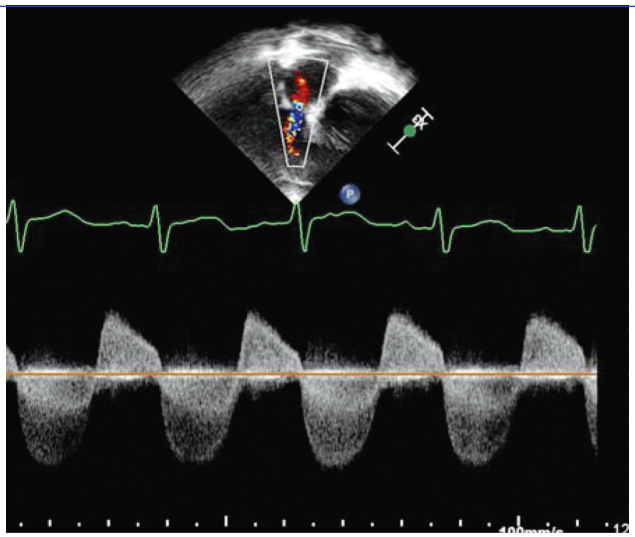
[Table/Fig-2]: Electrocardiogram reveals right axis deviation, RV hypertrophy, and right atrial enlargement.



[Table/Fig-3]: Echo shows VSD of 10 mm, overriding aorta, RV hypertrophy.



[Table/Fig-4]: Echo shows absent valve tissue.



[Table/Fig-5]: Echo shows "to-and-fro" flow in dilated pulmonary arteries.

Case 2

Adolescent TOF with Pulmonary Atresia: A 14-year-old male presented with progressive dyspnoea on exertion. He was born by normal vaginal delivery after an uneventful perinatal period. At four months of age, during evaluation for a lower respiratory infection, he was incidentally found to have a congenital heart disease and advised surgery, but surgery was deferred due to socioeconomic constraints. He remained on intermittent follow-up with haemoglobin 18 g/dL, haematocrit 55%.

From seven years of age, he developed dyspnoea on exertion corresponding to NYHA class II, gradually progressing to class III over the years. He became breathless while playing and had to stop and sit down. He noticed bluish discoloration of the tongue, particularly on exertion, along with headache and conjunctival suffusion. He also complained of right hypochondrial pain on exertion, which was relieved by rest. There was no history of orthopnoea, chest pain, palpitations, giddiness, pedal oedema, recurrent lower respiratory infections, joint pain, rash or rheumatic symptoms.

On examination, he was conscious, oriented, and afebrile. Central cyanosis and grade 2 clubbing were present. There was no pallor, icterus, pedal oedema or lymphadenopathy. Pulse was 70/min, regular, low volume, with no vessel wall thickening and no radio-radial or radio-femoral delay. Blood pressure was 100/60 mmHg in the right upper limb and 120/65 mmHg in the right lower limb. Oxygen saturation was 78% in the right upper limb, 79% in the right lower limb, and 81% in the left upper limb. Jugular venous pressure was not elevated.

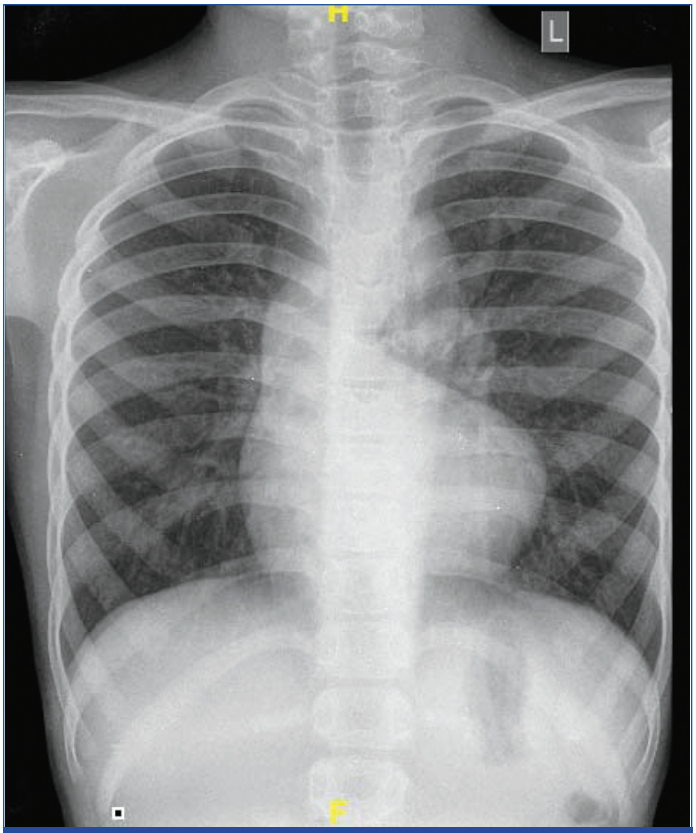
The chest was symmetrical, and the trachea was central. The apical impulse was situated in the fifth left intercostal space, 1 cm medial to the mid-clavicular line, with no specific character. A precordial bulge was present. RV impulse was palpable in the epigastrium with the tip of the finger, and there was a grade I parasternal heave. There was no palpable P2, no thrill, and no scars, sinuses or visible precordial pulsations.

On percussion, the left heart border corresponded to the apical impulse and the right heart border to the right sternal border. Liver dullness was felt in the right fifth intercostal space and Traube's space was resonant. Auscultation revealed normal S1 and S2 at the mitral, tricuspid and aortic areas. At the pulmonary area, there was a single loud S2 with absent P2. No murmurs or added sounds were heard.

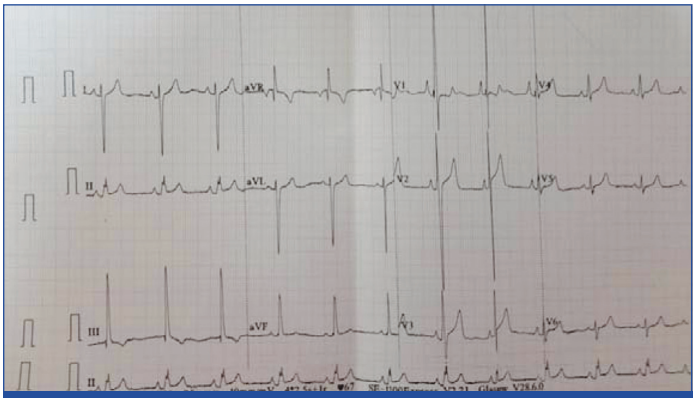
The working diagnosis was congenital cyanotic heart disease, TOF with pulmonary atresia, in failure, NYHA class III at the time of evaluation, with mild ventricular dysfunction due to severe chronic volume overload on the Left Ventricle (LV) due to excessive flow through large, non-restrictive Major Aortopulmonary Collateral Arteries (MAPCAs) that return blood to the left atrium increasing LV workload, sinus rhythm and no evidence of infective endocarditis. He had been on propranolol for 13 years and a diuretic for the preceding three months. Further evaluation with chest radiography [Table/Fig-6], electrocardiogram [Table/Fig-7], and echocardiogram [Table/Fig-8,9] confirmed the diagnosis of TOF with pulmonary atresia. A cath study was done, which confirmed large unrestricted MAPCAs, showed pulmonary venous hypertension, and pulmonary arterial anatomy and systemic-to-pulmonary collateral was defined. Definitive surgical repair with RV to pulmonary artery conduit with unifocalisation of MAPCAs planned.

Case 3

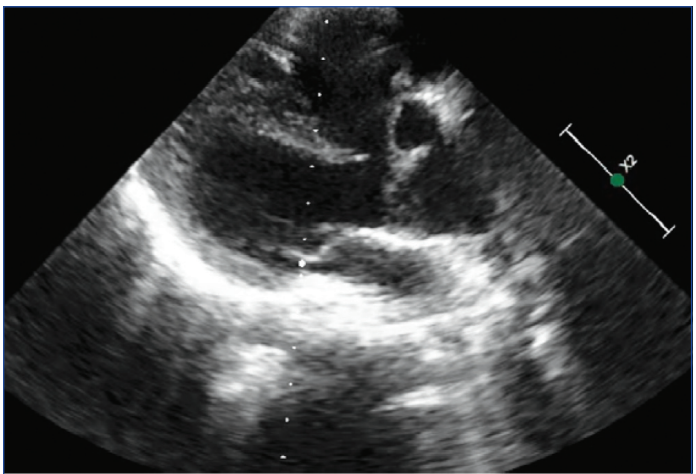
Adult TOF with Pulmonary Atresia After Bilateral BT Shunts: A 31-year-old woman, educated to the school level, presented with dyspnoea on exertion for one year, gradually progressing to New York Heart Association (NYHA) class II. She experienced breathlessness while climbing stairs and walking for more than five minutes. There was no orthopnoea or paroxysmal nocturnal dyspnoea. She reported intermittent pedal oedema that improved with medication. There



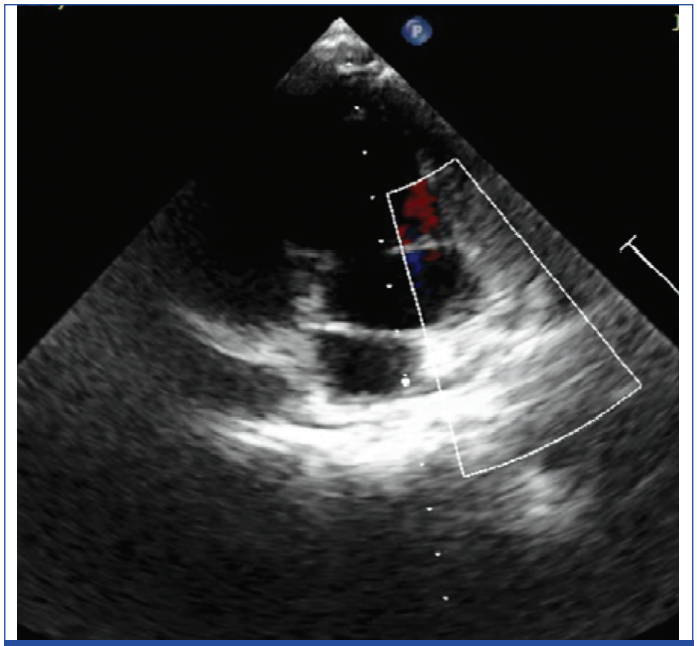
[Table/Fig-6]: X-ray shows decreased (oligemic) lung fields, a prominent aorta and an "empty" or concave pulmonary artery segment (pulmonary bay) due to the blocked connection to the lungs, with blood flow coming from Major Aortopulmonary Collateral Arteries (MAPCAs). The classic "boot-shaped" heart (RV hypertrophy) is less common here than in standard TOF, as the RV doesn't pump much to the lungs.



[Table/Fig-7]: Electrocardiogram shows RV Hypertrophy (RVH), Right Axis Deviation (RAD), and often signs of a Right Bundle Branch Block (RBBB), with large R waves in V1 and S waves in lateral leads, reflecting the overloaded RV.



[Table/Fig-8]: Echo shows a large Ventricular Septal Defect (VSD) 9 mm, an overriding aorta, and a thickened RV.



[Table/Fig-9]: Echocardiogram shows no connection (atresia) from the RV to the pulmonary artery, with pulmonary blood flow often coming from a Patent Ductus Arteriosus (PDA) or Major Aortopulmonary Collateral Arteries (MAPCAs).

was no history of right upper abdominal pain, haemoptysis, fever, weakness, blurring of vision, conjunctival suffusion, headache, or leg ulcers with haemoglobin 17 g/dL and haematocrit 58%.

She had been told she had a heart disorder at birth, though details were unavailable. She remained asymptomatic until five years of age, when she developed exertional dyspnoea during play, relieved by squatting. At 13 years, she began to experience giddiness, which became more frequent. At 15 years, she noticed bluish discolouration of her fingertips and lips on exertion, which worsened over time. She underwent a palliative classic BT shunt surgery at 15 years, after which her cyanosis improved. Three years later, at 18 years, her symptoms recurred, her palliative shunt got occluded, and she underwent a second palliative procedure on the contralateral side of the chest. Following the second surgery, cyanosis disappeared, but she started having recurrent lower respiratory tract infections, which were not present previously. Definitive repair may have been delayed due to socioeconomic reasons and lack of awareness, and she stopped going for follow-up after the second palliative surgery. She remained relatively well till the age of 30 years, after which she started becoming dyspnoeic.

There was no history of diabetes, hypertension, bronchial asthma, tuberculosis, or seizures. She followed a mixed diet, had normal bowel habits, regular 3/30 menstrual cycles without clots, and no relevant family history. She was taking sildenafil to "reduce pressures in the heart" for the past six months.

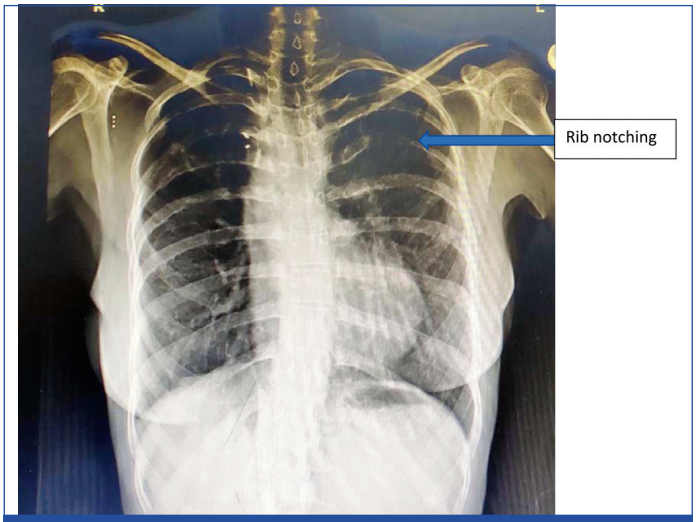
On examination, she was conscious, oriented, and afebrile. There was no pallor, pedal oedema, or lymphadenopathy. There was mild cyanosis, which was not very apparent due to dark skin tone, and she had grade 3 pandigital clubbing. Pulse was 85/min, low volume, with no special character, vessel wall thickening, or radio-radial or radio-femoral delay. Blood pressure was 102/72 mmHg in the right upper limb and 102/82 mmHg in the right lower limb. Peripheral oxygen saturation was around 88-90% in the upper and lower limbs. Jugular venous pressure was elevated 10 cm above the sternal angle with prominent a waves suggesting RV dysfunction.

The chest was symmetrical with the trachea in the midline. The apical impulse was felt in the fifth left intercostal space, 1 cm medial to the mid-clavicular line, and had no specific character. There was no precordial bulge, parasternal heave, palpable P2, or thrill. Bilateral posterior thoracic scars were present, consistent with prior BT shunts. On percussion, the right heart border corresponded to the right sternal border and the left heart border to the apical

impulse. Liver dullness was felt in the right fifth intercostal space, and the left second intercostal space was resonant.

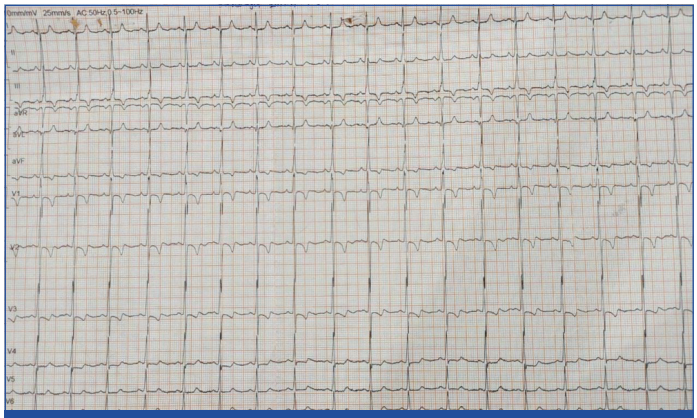
On auscultation, S1 and S2 were heard in all areas. In the pulmonary area, S2 was single with loud A2. A grade 3/6 continuous murmur was heard over the pulmonary area and in the back, suggestive of a functioning systemic-to-pulmonary shunt. There were no additional murmurs or signs of active endocarditis.

Chest X-ray Posteroanterior view [Table/Fig-10] revealed the presence of right sided aortic arch, RV type of apex, boot-shaped heart, no cardiomegaly with absent pulmonary arteries. Rib notching was present in the inferior surface of the medial aspect of the 2nd and 3rd intercostal ribs on both sides, correlating with the MAPCAs. Erosion of the third intercostal rib was also present. There were patchy areas of hypoperfusion in bilateral upper lobes with normal perfusion in bilateral lower lobes corresponding to areas perfused by modified BT shunt and MAPCAs.



[Table/Fig-10]: X-ray shows increased pulmonary blood from the BT shunt, boot-shaped heart, and surgical clips or coils near the shunt site, confirming its placement and function. Rib notching was present in the inferior surface of the medial aspect of the 2nd and 3rd intercostal ribs on both sides, correlating with the MAPCAs.

Electrocardiogram [Table/Fig-11] and echocardiogram [Table/Fig-12,13] showed features of TOF with pulmonary atresia, NYHA class II, status post bilateral BT shunts with RV dysfunction with Tricuspid Annular Plane Systolic Excursion (TAPSE) of 12 mm.

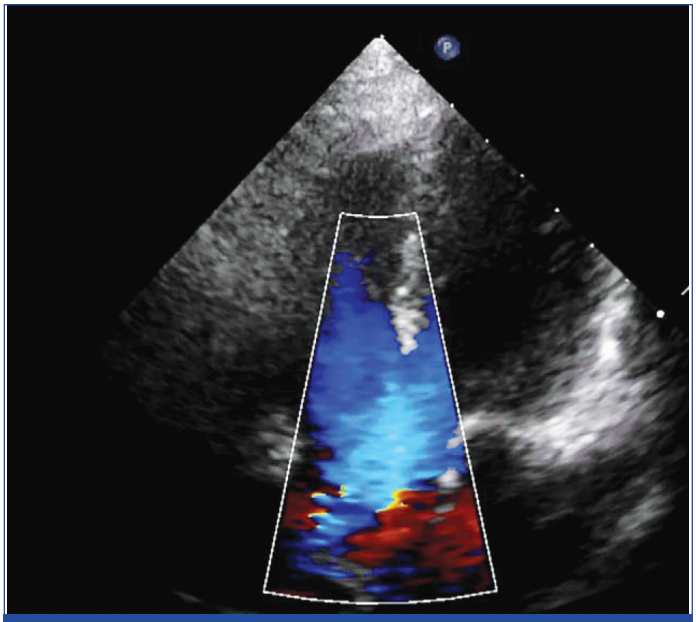


[Table/Fig-11]: Electrocardiogram shows right axis deviation and RV hypertrophy.

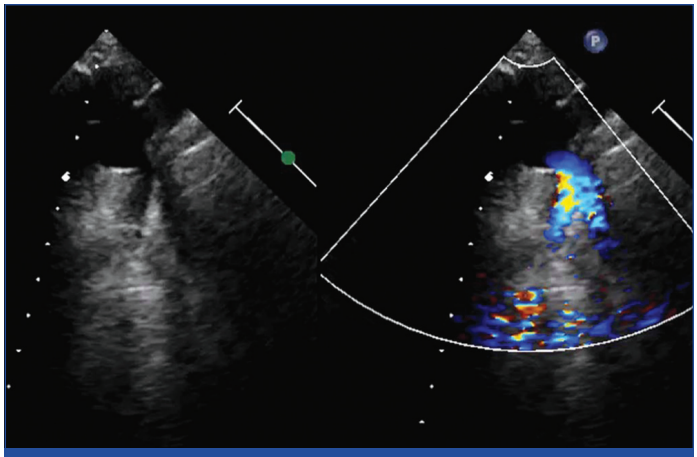
A cath study was done to assess pulmonary artery size, shunt patency and collateral circulation and to determine the feasibility of completion repair versus further palliative intervention and was planned for unifocalisation of MAPCAs.

DISCUSSION

The TOF with RVOT variants shows marked heterogeneity in anatomy, pulmonary blood flow, and clinical course, leading to age-specific patterns of presentation [1]. Our three cases demonstrate this



[Table/Fig-12]: Echocardiogram shows a patent Ventricular Septal Defect (VSD) with overriding aorta.



[Table/Fig-13]: Echocardiogram shows the shunt itself (flow from subclavian to pulmonary artery) and a small or absent native pulmonary artery system.

spectrum: an acyanotic infant with TOF and APV dominated by respiratory symptoms, an adolescent with unoperated TOF with pulmonary atresia and severe cyanosis, and an adult survivor with TOF/pulmonary atresia palliated by bilateral BT shunts [Table/Fig-14].

In the first case of TOF with APV, the seven-month-old male presented acyanotic with respiratory distress from recurrent infections and wheezing due to massively dilated pulmonary arteries compressing bronchi, as confirmed by chest X-ray showing aneurysmal central pulmonary arteries and echocardiography revealing absent valve tissue with to-and-fro flow [2]. This APV Syndrome (APVS), occurring in 3-6% of TOF cases, leads to free pulmonary regurgitation and aneurysmal dilatation, distinguishing it from classic TOF's boot-shaped heart [3]. Treatment involved medical management with antibiotics and nebulisation alongside surgical Ventricular Septal

TOF variant	Presentation	Initial Palliation	Possible definitive repair
Case 1 Absent Pulmonary Valve (APV) (Infant)	Respiratory distress, no cyanosis	Antibiotics, ventilation support if needed	VSD closure + RVOT patch + PA plasty
Case 2 Pulmonary Atresia (Adolescent, unoperated)	Cyanosis, polycythaemia	Beta-blockers, phlebotomy if needed	RV-PA conduit + VSD Closure + MAPCA unifocalisation
Case 3 Pulmonary Atresia (Adult, post-BT)	Dyspnoea, RV failure	Sildenafil, diuretics	Unifocalisation + VSD closure

[Table/Fig-14]: Illustrating the presentation and management of the three cases.
VSD -Ventricular Septal Defect

Defect (VSD) closure, RVOT reconstruction via transannular patch, and pulmonary artery reduction plasty to relieve airway compression, aligning with recommendations for early intervention in infants to prevent tracheobronchomalacia [2,4].

In the second case, a 14-year-old male with TOF and pulmonary atresia exhibited progressive cyanosis (SpO_2 78-81%), polycythaemia (Hb 18 g/dL), and NYHA class III dyspnoea due to untreated pulmonary atresia (10-15% of TOF), reliant on unrestricted MAPCAs causing left ventricular overload and pulmonary venous hypertension, evidenced by oligemic lung fields on X-ray and cath confirmation [5]. Long-term propranolol and diuretics managed symptoms, however, delayed repair due to socioeconomic barriers highlights challenges in low-resource settings [1]. Planned definitive repair includes RV-to-Pulmonary Artery (PA) conduit with MAPCA unifocalisation, guided by intraoperative flow studies to assess post-repair pulmonary pressures, as this predicts feasibility and reduces risks like high RV pressure [6].

In the third case with TOF with pulmonary atresia with palliative shunts, the 31-year-old female showed NYHA class II dyspnoea, RV dysfunction (TAPSE 12 mm), elevated Jugular Venous Pressure (JVP), and patent shunts (continuous murmur, SpO_2 88-90%) after classic BT shunts at ages 15 and 18, complicated by post-shunt infections and rib notching from MAPCAs on X-ray [5]. Bilateral modified BT shunts provided palliation by augmenting pulmonary flow via subclavian-to-PA anastomosis but risk over circulation, shunt occlusion, and long-term RV strain requiring sildenafil [7]. Cath-guided unifocalisation and complete repair are planned, emphasising lifelong surveillance for arrhythmias and dysfunction in adult survivors [8]. Early diagnosis via echocardiography and tailored palliation (e.g., BT shunts) bridge to complete repair, improving survival beyond infancy despite late presentations [1,9].

CONCLUSION(S)

These cases illustrate the age-specific clinical spectrum of TOF variants. Management priorities differ across age groups: airway protection in infants with APV syndrome, prevention of complications from chronic cyanosis in adolescents, and long-term surveillance for RV dysfunction and arrhythmias in adults with prior palliation. In resource-limited settings, multidisciplinary care with cath mapping optimises outcomes, reducing complications like RV dysfunction (common in 20-30% of adults). Future strategies may incorporate advanced imaging for prenatal counselling. These findings emphasise the need for heightened awareness, advanced imaging, and socioeconomic support to facilitate timely diagnosis and definitive management, ultimately mitigating long-term complications and improving survival in TOF variants.

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