Incidental finding of supra cardiac total anomalous pulmonary venous connection with Tetralogy of Fallot: A case report

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Abstract

Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disorder (CHD) worldwide and is known to be associated with a variety of other CHDs. This case report explores a rare incidental combination of TOF and supracardiac total anomalous pulmonary venous connection (TAPVC). Such a coexistence is exceedingly uncommon with only a handful of reported cases which prompts the need for documentation, especially its management in a developing country. Prompt diagnosis is challenging due to atypical symptoms which emphasises the importance of comprehensive imaging. Surgical intervention, guided by pre-operative cardiac catheterisation, addressed the anatomical complexities and facilitated successful single-stage repair. The patient was discharged on the fifth post-operative day. Follow-up echocardiogram demonstrated no residual ventricular septal defect, unobstructed right ventricular outflow, normal biventricular function, and unrestricted pulmonary venous drainage to the left atrium.

Keywords: Congenital heart defect, Tetralogy of Fallot, TAPVC.

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Introduction

TOF is the most common cause of cyanotic CHD and earliest documented cardiac lesion. A survey conducted in 2015 reports the prevalence of CHD as 3.4 per 1,000 live births in the rural communities of Pakistan.[1] It is commonly associated with other defects such as an atrial septal defect (ASD), patent ductus arteriosus (PDA), and anomalous coronary arteries.

TAPVC is defined as an abnormality in which the pulmonary veins drain into the systemic vasculature rather than the left atrium of the heart, resulting in no systemic circulation of blood. The association of TOF with TAPVC is considerably rare, with only a handful of cases reported in the past years. This report describes a two-and-a-half-year-old boy presenting with respiratory distress and cyanosis. He was diagnosed with TOF at birth and further investigation revealed that he had associated supracardiac TAPVC.

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Case Report

A two-and-a-half-year-old boy, weighing 10 kg, presented with escalating cyanosis at Ittefaq Hospital [Trust], Lahore on October 27, 2023. The parents reported intermittent bluish discolouration of lips and extremities since five months, that intensified during crying or feeding. Initially diagnosed with TOF and scheduled for elective surgery at one year of age, he remained asymptomatic with mild cyanosis until worsening at age 2.5 years. Initial echocardiogram revealed TOF findings but missed anomalous venous return.

During pre-operative workup, chest radiograph showed a peculiarly wide mediastinum. A careful review of the echocardiogram revealed increased superior vena cava (SVC) flow, leading to the discovery of a large vertical vein redirecting pulmonary venous return to the right atrium

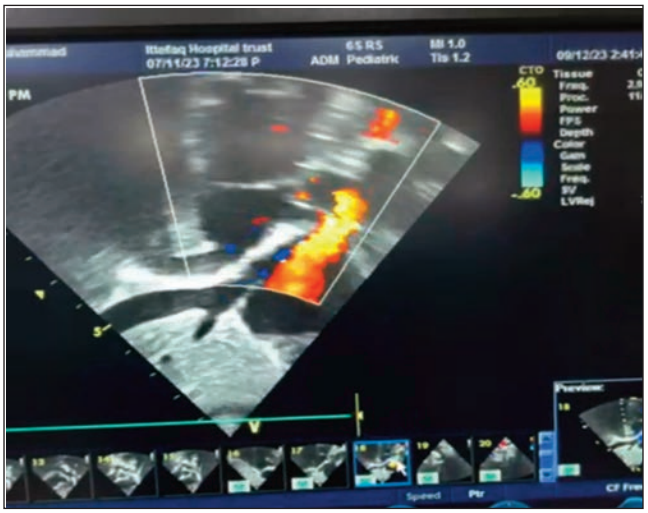

Figure-1: Pre-operative echocardiography in bi-caval view showing increased venous return into the Superior Vena Cava.

(RA) via dilated innominate and superior caval veins (Figures 1, 2). Diagnostic cardiac catheterisation confirmed unobstructed supracardiac TAPVC/D (Figure 3) with low mean pulmonary artery pressures and adequate left ventricular (LV) volume, alongside a moderate unrestricted atrial septal defect (ASD) (Figure 4).

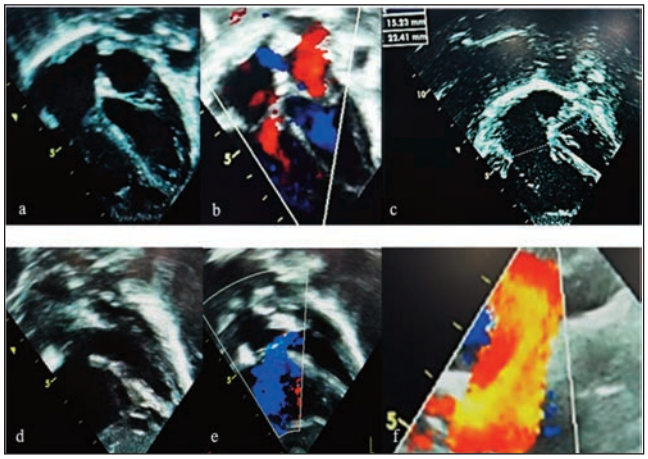

Figure-2: Pre-operative echocardiography; a, b, c: balanced ventricles with good size Left Ventricle; d, e: single large subaortic Ventricular Septal Defect; f: vertical vein draining pulmonary venous return to Superior Vena Cava.

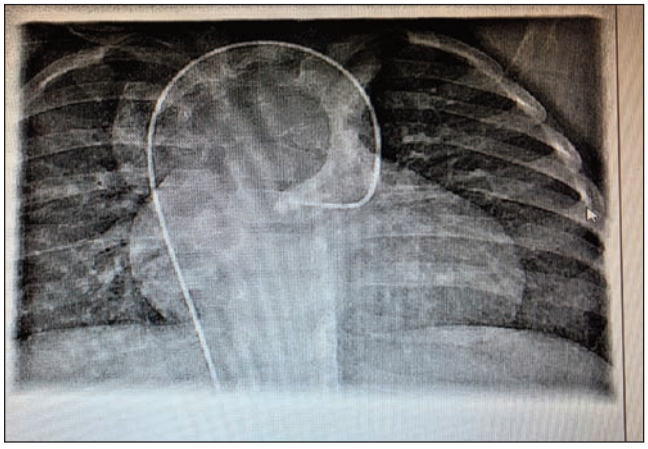

Figure-3: Angiogram showing venous injection passing from Inferior Vena Cava to Superior Vena Cava into the innominate vein, passing through the vertical vein and into the common pulmonary confluence.

Figure-4: Right ventricular outflow angiogram showing a: infundibular and valvular PS; b: (Levophase) total pulmonary venous return to RA via vertical vein, innominate vein and Superior Vena Cava.

Standard median sternotomy was done, and pericardial patch was harvested. Cardiopulmonary bypass (CPB) was established with aortic and bicaval cannulation at moderate hypothermia of 30°C. The vertical vein was dissected and looped. Common pulmonary confluence was also dissected after dividing pericardial reflections. The aorta was cross clamped after performing antegrade del Nido cardioplegia.

The RA was opened transversely, extending the incision through the interatrial septum to the left atrial posterior wall and towards the left atrial appendage. Identification of the ventricular septal defect (VSD) followed examination of the right ventricular outflow through the tricuspid valve. Opening the main pulmonary artery (MPA) facilitated trans-atrial and trans-pulmonary resection of the obstructing right ventricular bundles, and closure of the VSD with a Dacron patch. The common pulmonary confluence was broadly opened and anastomosed to the exposed left atrial posterior wall. ASD and MPA patch plasty were done with the pericardial patch. The RA was closed, de-airing was done, and the aorta cross clamp was removed. The patient weaned off CPB with normal sinus rhythm with good haemodynamic. Peri-operative epicardial echocardiography confirmed no residual VSD, unobstructed right ventricular outflow, trivial pulmonary regurgitation, and unrestricted pulmonary venous drainage to the left atrium. Haemostasis was secured, atrial and ventricular pacing leads and mediastinal drains were placed, and standard closure was done.

The post-operative course was uneventful, with extubation and weaning off inotropes on the first post-operative day, transfer out of the ICU on the second day, and discharge on the fifth post-operative day. Follow-up echocardiogram demonstrated no residual VSD, unobstructed right ventricular outflow, normal biventricular function, and unrestricted pulmonary venous drainage to the left atrium.

Discussion

TOF has a multifactorial aetiology, developing interventricular septal malalignment anteriorly, leading to the development of outlet VSD, overriding aorta, and right ventricular (RV) infundibular narrowing with subsequent RV hypertrophy. Common variations include TOF with pulmonary atresia, TOF with absent pulmonary valve syndrome, and TOF with complete atrioventricular septal defect. Additional associations often include ASD, anomalous coronary arteries, additional muscular VSDs,

straddling tricuspid valve, and right aortic arch. Anomalous pulmonary drainage, particularly partial or total anomalous pulmonary venous connection, has been reported sporadically. Though antenatal diagnosis is common, undiagnosed cases typically manifest around the second month of life, presenting with worsening cyanosis and hyper-cyanotic spells at four to six months.[2]

TAPVC/D is an uncommon congenital heart defect (up to 3%) and is classified based on the route of drainage (supracardiac, cardiac, infracardiac, and mixed). The cause remains unclear, though it is associated with abnormal drainage of the pulmonary venous confluence to the RA due to the failure of pulmonary and cardiac mesoderms during the development of the left atrium. Presentation occurs at birth with symptoms like cyanosis, tachypnoea, feeding interruptions, and poor weight gain, necessitating early surgical intervention.[3]

Supracardiac TAPVC/D is extremely rare in conjunction with TOF, with only a few reported cases in literature.[4] Due to the absence of typical signs and symptoms, TAPVC/D may be overlooked, especially when pulmonary venous return is unobstructed. Thorough evaluation through chest radiography and focussed echocardiography is crucial to prevent intra-operative surprises.[5] Concerns associated with this condition include anatomical variations, potential obstruction in venous channels, high pulmonary artery pressure, and a relatively small left ventricle (LV) due to chronic low-volume venous return.[4]

Pre-operative cardiac catheterisation helps confirm venous channel connections, assess potential obstructions, and determine pulmonary pressures. The LV size has been a cause of concern, and data shows that single-stage repair is feasible for unobstructed varieties with sufficient blood flow to the LV as was in the current case.[3] In some case reports a two-stage repair has been used due to concerns about chronic under-filling and insufficient support for systemic circulation.[6]

Conclusion

TOF with TAPVC/D provides a rare combination of CHDs requiring special pre-operative and peri-operative consideration. Pre-operative diagnosis based on carefully planned investigations helps alleviate various anatomical and haemodynamic concerns leading to a good surgical outcome.

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Author Contribution:
MUK: Literature search, discussing presentation with patient's family and writing.
AUQ & MR: Confirmed diagnosis via echocardiography and discussing presentation with patient's family.
MAK: Discussing presentation with patient's family, literature search and performed surgical repair.