Case study

Tetralogy of Fallot with Total Anomalous Pulmonary Venous Connection: a rare variant

ABSTRACT

Tetralogy of Fallot (TOF) with total anomalous pulmonary venous connection (TAPVC)is a rare entity involving ~~the~~ disordered development of arterial and venous ends of the heart. Clinical manifestations are similar to classical TOF with masked findings of TAPVC, thus posing challenge to clinicians. Preoperative diagnosis is mandatory as repair of both defects is required simultaneously. Palliative modified Blalock-Taussig shunt alone is contraindicated as it may lead to catastrophic complications.

Comment [SD1]: Need to be more specific. This is a very vague statement.

Comment [SD2]: This is not necessary in abstract. Keep only in discussion.

Keywords: Tetralogy of Fallot, Total Anomalous Pulmonary Venous Connection, Modified Blalock-Taussig Shunt, Total Intracardiac Repair, Rerouting of PulmonaryVeins.

Comment [SD3]: This paper does not address these terms, so better not use in the keywords

1. INTRODUCTION

Tetralogy of Fallot (TOF) with total anomalous pulmonary venous connection (TAPVC) is a distinctly rare congenital anomaly with important clinical implications in diagnosis as well as in management [1], [2].Diagnosis is completely missed clinically as manifestations of TAPVC are masked due to right ventricular outflow tract obstruction (RVOTO) and is frequently missed in echocardiography. Certain clues raise suspicion of TAPVC in echocardiography. Subsequent work up may include CT, MRI or cardiac catheterization. Preoperative diagnosis is mandatory as simultaneous correction of TAPVC and TOF is required to avoid the complications. The purpose of this article is: (1) to add to the existing literature ~~to augment the reported volume~~ of this clearly uncommon congenital anomaly; (2) to illustrate how to suspect and diagnose the TAPVC associated with TOF; (3) to discuss the technical challenges in surgical correction imparted by anomalous route of pulmonary venous drainage &/or small sized left sided cardiac chambers.

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2. PRESENTATION OF CASE

An 11-month- old- girl child was brought to our hospital with history of bluish discoloration of fingers, toes and mucous membrane since birth in absence of significant history of feeding difficulty, recurrent lower respiratory tract infections, cyanotic spells or squatting. There was no history of similar illness or unexplained premature death in any of family members. Upon examination central cyanosis and grade 2 pan digital clubbing was detected. In ambient air her Oxygen saturation was 85% ~~with pulse oximeter~~. There was no precordial hyperactivity with single second heart sound, grade II/VI ejection systolic murmur in left second - third intercostal spaces and continuous murmur in left infra clavicular region.

Her electrocardiogram (ECG) revealed right ventricular hypertrophy with right axis deviation. Chest X-ray revealed boot shaped heart. ~~2 D E~~Two dimensional echocardiography revealed normal situs and levocardia. There was large ventricular septal defect (VSD) with aortic override of around 50% (figure 1) and significant RVOTO (maximum gradient of 68 mm of Hg predominantly at valvular level) (figure 2). There was right ventricular hypertrophy with right to left shunt across VSD (video 1). Pulmonary veins were opening into a common chamber located behind the left atrium (LA) which was draining into right atrium (video 2,3). LA was small with 8 mm ostium secundum atrial septal defect (OS ASD) with right to left shunt. Great arteries were normally related. Main pulmonary artery (10.3 mm) and its confluent branches were of adequate size. A small (2 mm) patent ductus arteriosus (PDA) was shunting systemic to pulmonary circulation as seen infigure 2 and video 4. The aortic arch was left sided.

Hence, the diagnosis of TOF with cardiac type TAPVC was ~~drawn~~ made which was subsequently confirmed with cardiac CT (figure 3) which nicely showed drainage of all the four pulmonary veins into the common chamber located behind the LA and below the bifurcation of pulmonary trunk. This common chamber was connecting to the right atrium without any obstruction in pulmonary venous drainage. No major aorto-pulmonary collaterals were visualized. Catheterization study was not performed as cardiac anatomy was well delineated with echocardiography and cardiac CT.

Oral Iron and beta blockers were started. Patient was referred to cardiothoracic surgery department with plan for early elective surgery in the form of total intracardiac repair along with rerouting the pulmonary venous drainage to LA.

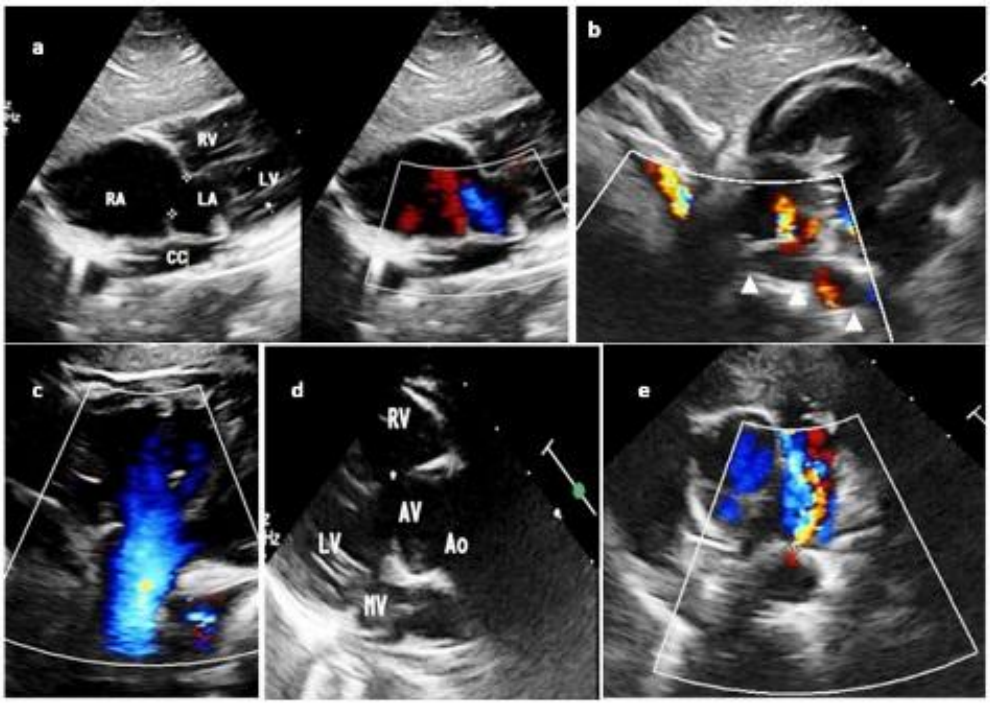

Figure 1. (a) Echocardiography in subcostal four chamber view demonstrated huge right atrium (RA), small left atrium (LA) with 8 mm ostium secundum atrial septal defect shunting right to left. Behind the left atrium is common chamber (CC). (b) Common chamber is (white arrow heads) draining into RA. (c & d) Five chamber and long axis views showing large ventricular septal defect shunting right to left with aortic (Ao) override. (e) Tiny patent ductus arteriosus (PDA) with shunt direction from aorta to pulmonary artery. LV- left ventricle, RV- right ventricle, MV- mitral valve, * nonrestrictive VSD.

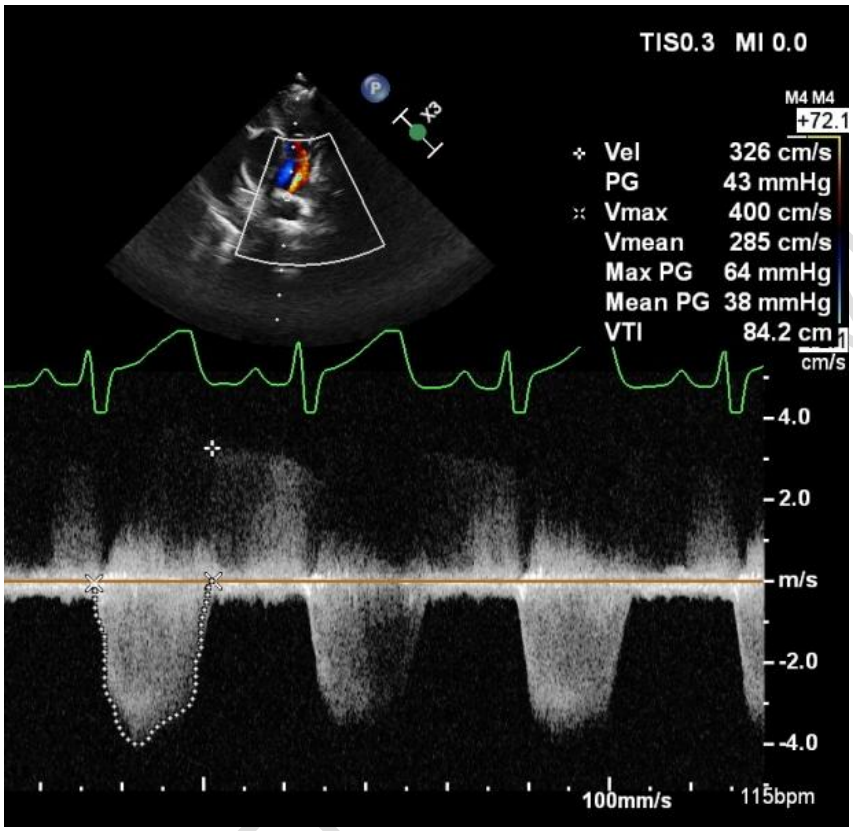

Figure 2. Continuous wave Doppler across right ventricular outflow showed significant gradient with the spectral wave pattern suggestive of valvular obstruction. Also the faint spectral wave of continuous flow across PDA was captured with significant pressure difference. PDA- patent ductus arteriosus

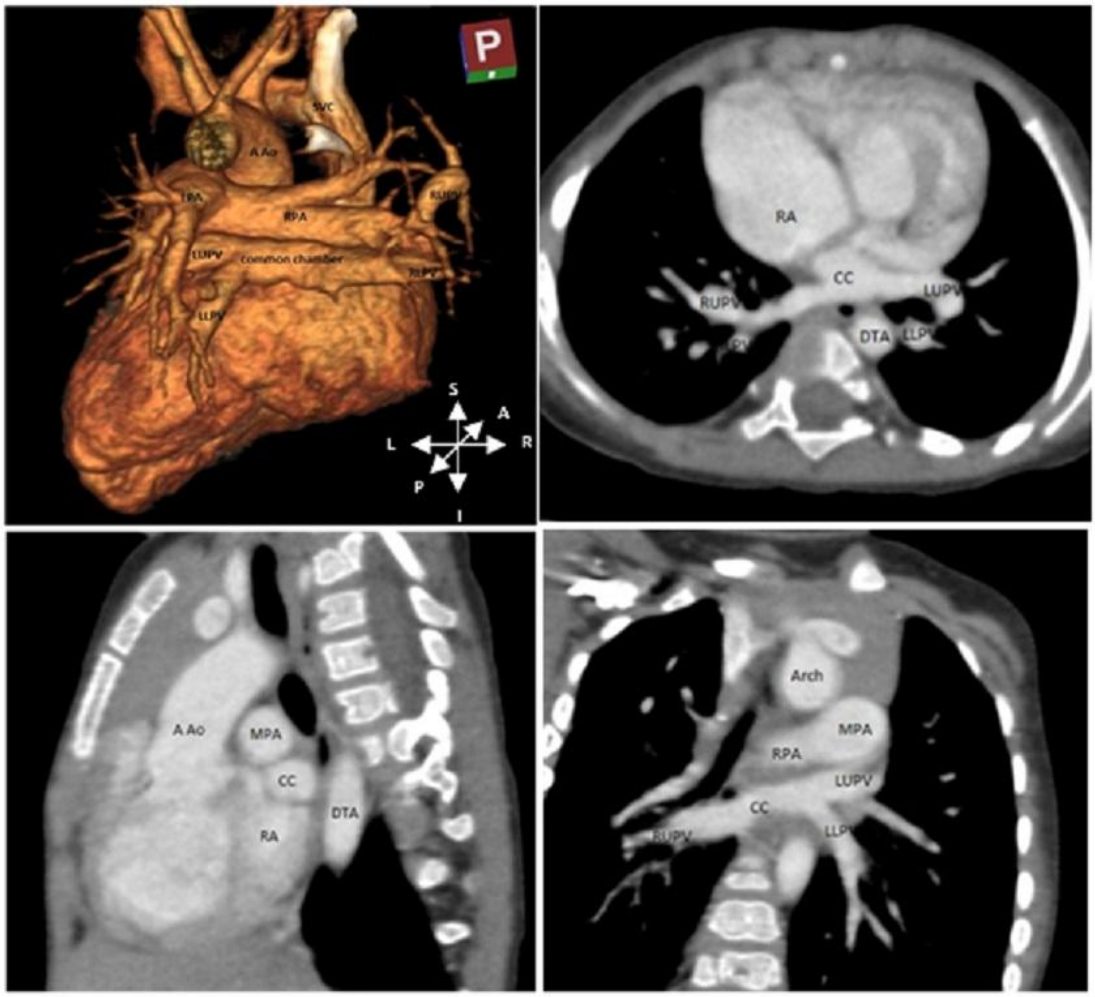

Figure 3. In clock wise order from left upper corner, volume rendered computed tomography image, transverse, coronal and sagittal sections revealing formation of common chamber (CC) by all four pulmonary veins behind small left atrium (LA) and below the pulmonary artery branches. This chamber in turn drains in to dilated right atrium (RA).A Ao- ascending aorta, Arch- aortic arch, DTA- descending thoracic aorta, LLPV- left lower pulmonary vein, LUPV- left upper pulmonary vein, RLPV- right lower pulmonary vein, RUPV- right upper pulmonary vein, SVC- superior vena cava.

3. DISCUSSION

Reported incidence of TOF with TAPVC is 0.26%of all the TOF cases [1]. Because of the distinctive rarity of this combination and clinical findings of classical TOF only, diagnosis is hardly ever clinched clinically and frequentlymissed in echocardiography, particularly in cardiac type TAPVC associated with large atrial septal defect (ASD)[2]. Not infrequently, this associated TAPVC is first documented when CT or MRI is performed during further work up [3], [4], [5].There is

significant contribution of the second heart field located in the pharyngeal mesoderm, in the development of arterial and venous ends of the heart. Disordered development involving both these ends leads to co-occurrence of TOF with TAPVC[6], [7].

Possible clues to suspect anomalous pulmonary venous drainage in echocardiography include (a) large RA coupled with small LA; (b) presence of shelf like partition in LA giving an initial impression of cor triatriatum; (c) visualization of another chamber behind the LA not communicating with it; (d) enlarged coronary sinus in absence of left sided superior vena cava (LSVC) and (e) demonstration of vertical vein in suprasternal view.

It is of paramount ~~important~~ importanceto have a correct preoperative diagnosis because simultaneous complete repair of TOF and TAPVC is mandatory. These patients present with classical signs and symptoms of TOF. On isolated clinical ground, presence of TAPVC can't be suspected as RVOTO masks the findings of TAPVC. Even obstructive TAPVC escapes diagnosis and remains concealed becausereduced pulmonary preload prevents development of pulmonary venous congestion. Treatment involves repair of TAPVC by anastomosing pulmonary venous confluence to the left atrium along with RVOTO and VSD repair if there are adequate sized cardiac chambers on both side. If left sided chambers are small, then correction of TAPVC with systemic- pulmonary shunt is performed in first stage followed by repair of TOF in second stage. Isolated palliative treatment in the form of systemic to pulmonary shunt is not done to avoid the development of pulmonary edema especially if there is obstructive variant of TAPVC. If the patient is unsuitable for complete correction, but requires palliative surgery to increase pulmonary blood flow, correction of the anomalous pulmonary venous return should be performed at the same time. In some of the patients there may be pulmonary vascular changes despite the RVOT obstruction and this may necessitate leaving or creating a small ASD by atrial balloon septostomy or surgical excision of the atrial septum to provide "atrial pop off" in the event of significantly raised right-sided pressures[5].

4. CONCLUSION

- TOF with TAPVC is a rare congenital cardiac anomaly.
- Diagnosis is usually missed clinically as clinical manifestations typically match to that of isolated classical TOF with concealed findings of TAPVC.
- First suspicion of TAPVC usually comes with certain clues in echocardiography. Subsequent work up in the form of CT, MRI or cardiac catheterization may establish the diagnosis.
- Preoperative diagnosis is mandatory as simultaneous correction of TAPVC and TOF is required to avoid the complications.

Comment [SD6]: Conclusion should reflect the work of your paper and not the theory related to the topic. Please re-write this concisely.

7. CONSENT

Written informed consent was obtained from the patients for publication of this case report and any accompanying images.

8. REFERENCES

Comment [SD7]: Referencing numbering is not correct. There are empty numbers without referencing.

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13. [7]. Vergara P, Digilio MC, Zorzi AD, et al. Genetic Heterogeneity and Phenotypic Anomalies in children with Atrioventricular Canal Defect and Tetralogy of Fallot. Clin Dysmorphol 2006; 15:65-70.

LEGENDS

Videos

Comment [SD8]: Videos are not included in the manuscript.

Video 1. 2 D Echocardiography loop of subcostal five chamber view showing large ventricular septal defect shunting right to left with aortic override and right ventricular hypertrophy.

Video 2. 2 D Echocardiography with color Doppler in subcostal four chamber view showed huge right atrium, small left atrium and common chamber located behind left atrium.Common chamber is draining into right atrium.

Video 3. 2 D Echocardiography with color Doppler in subcostal sagittal view demonstrated common chamber draining into right atrium.

Video 4. A small patent ductus arteriosus was shunting systemic to pulmonary circulation.