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Tetralogy of Fallot with Total Anomalous Pulmonary Venous Connection: A Rare Variant

A. K. Sharma [a++*] and S. Khatri [a#]

[a] Department of Cardiology, Fortis Escorts Hospital, Jaipur, Rajasthan, India.

Authors' contributions

This work was carried out in collaboration between both authors. Both authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Tetralogy of Fallot with total anomalous pulmonary venous connection is a rare entity. Due to disordered development of arterial and venous ends of the heart there is anomalous venous return coupled with outflow tract anomalies. Clinical manifestations are similar to classical Tetralogy of Fallot with masked findings of anomalous pulmonary venous return, thus posing challenge to clinicians. Preoperative diagnosis is mandatory as repair of both defects is required simultaneously.

Keywords: Tetralogy of Fallot; total anomalous pulmonary venous connection; total intracardiac repair; rerouting of pulmonary veins.

[++]Consultant;
[#]Additional Director;
**Corresponding author: Email: ashwanikumarsharma00@gmail.com;*

1. INTRODUCTION

Tetralogy of Fallot (TOF) with total anomalous pulmonary venous connection (TAPVC) is a distinctly rare congenital anomaly with important clinical implications not only in diagnosis but also 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 cardiac tomography (CT), magnetic resonance imaging (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 to add to the existing literature of this rare congenital anomaly and to discuss the technical challenges in surgical correction imparted by anomalous route of pulmonary venous drainage &/or small sized left sided cardiac chambers.

2. PRESENTATION OF CASE

An 11-month- old- girl child was brought the 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. On examination central cyanosis and grade 2 pan digital clubbing was detected. In ambient air her Oxygen saturation was 85%. There was no precordial hyperactivity. Auscultation revealed single second heart sound with 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. Two-dimensional echocardiography revealed normal situs and levocardia. There was large ventricular septal defect (VSD) with aortic override of around

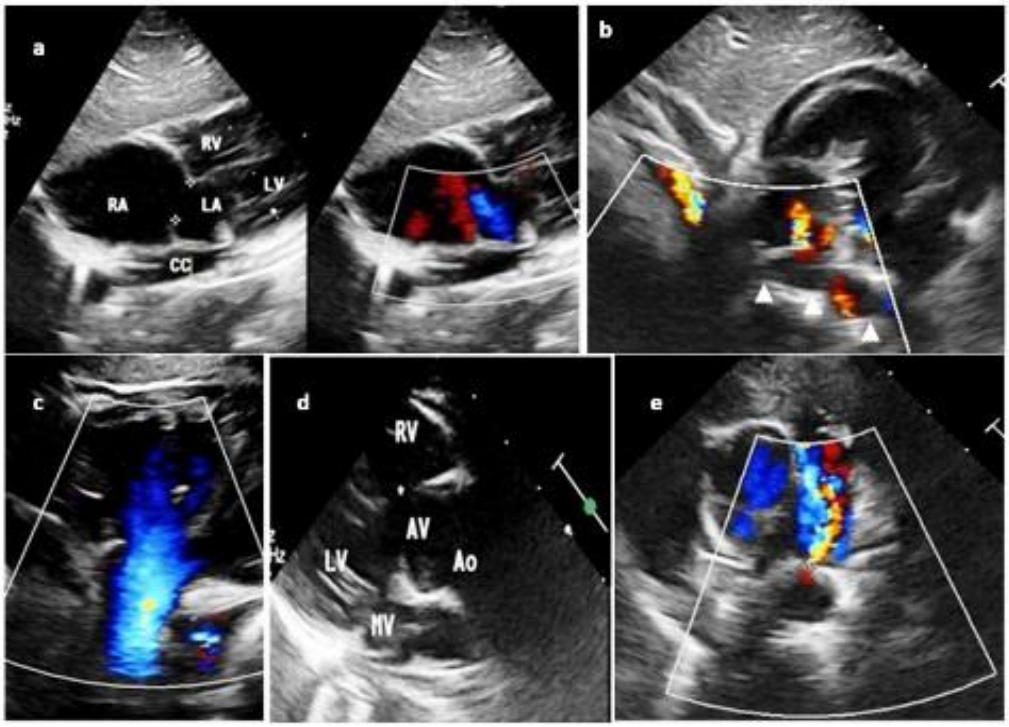

Fig. 1. (a) Echocardiography in subcostal four chamber view demonstrated huge right atrium (RA), small left atrium (LA) with 8 mm ostium secundum atrial septal defect shunting right to left. Behind the left atrium is common chamber (CC). (b) Common chamber is (white arrow heads) draining into RA. (c & 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*

50% (Fig. 1) and significant RVOTO (maximum gradient of 68 mm of Hg predominantly at valvular level) (Fig. 2). There was right ventricular hypertrophy with right to left shunt across VSD (Video 1). Pulmonary veins (PV) were opening into a common chamber located behind the left atrium (LA) which was draining into right atrium (RA) (Videos 2, 3). LA was small with 8 mm ostium secundum atrial septal defect (OS ASD) shunting right to left. Great arteries were normally related. Main pulmonary artery (10.3 mm) and its confluent branches were adequate in size. A small (2 mm) patent ductus arteriosus (PDA) was shunting systemic to pulmonary circulation (Fig. 2 and Video 4). The aortic arch was left sided.

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

The child underwent definitive surgery in the form rerouting pulmonary venous confluence to the posterior wall of LA along with RVOTO and VSD repair. She had uneventful post-operative course and was extubated on post- operative day 2. Intraoperative findings were consistent with pre-operative diagnosis. She is doing well at 2 year follow up.

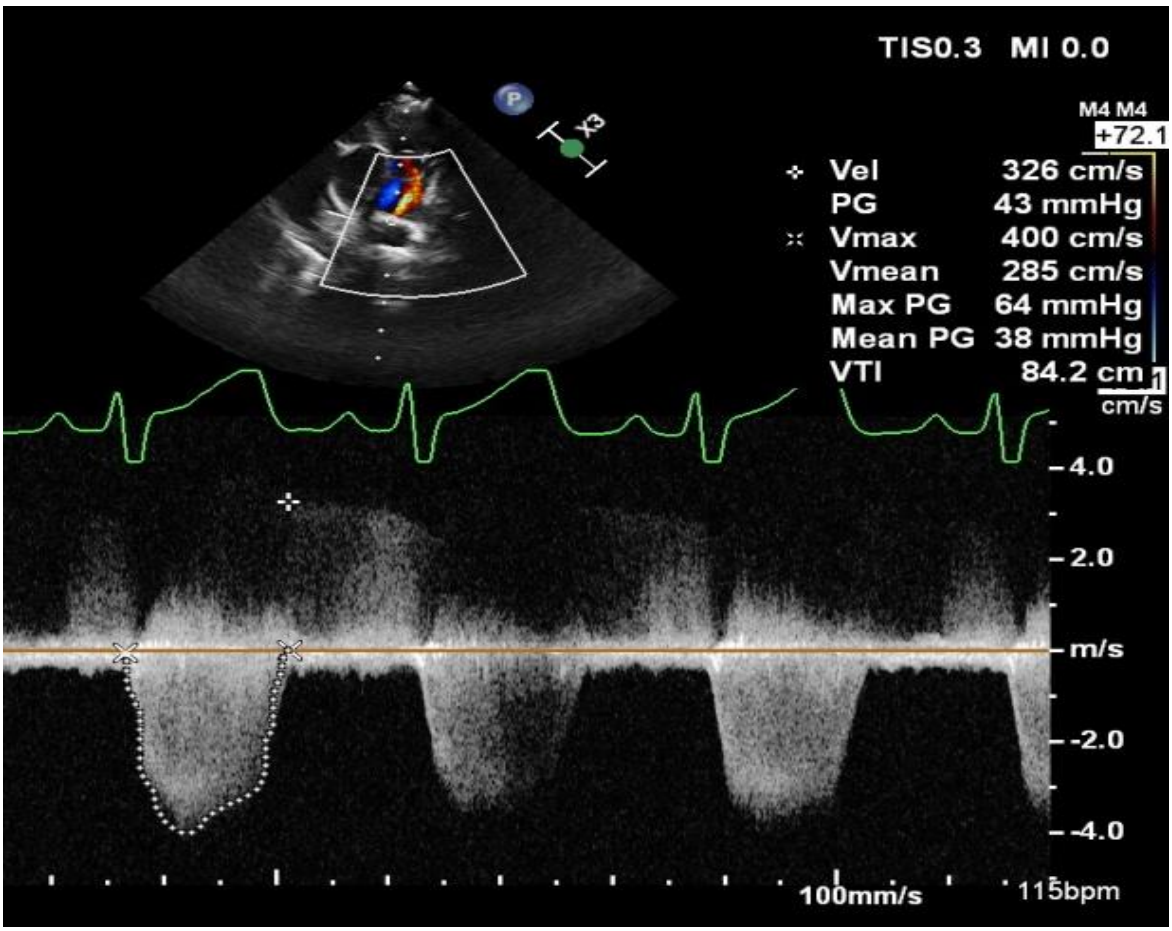

Fig. 2. Continuous wave Doppler across right ventricular outflow showed significant gradient with the spectral wave pattern suggestive of valvular obstruction. Also the faint spectral wave of continuous flow across PDA was captured with significant pressure difference

PDA- Patent Ductus Arteriosus

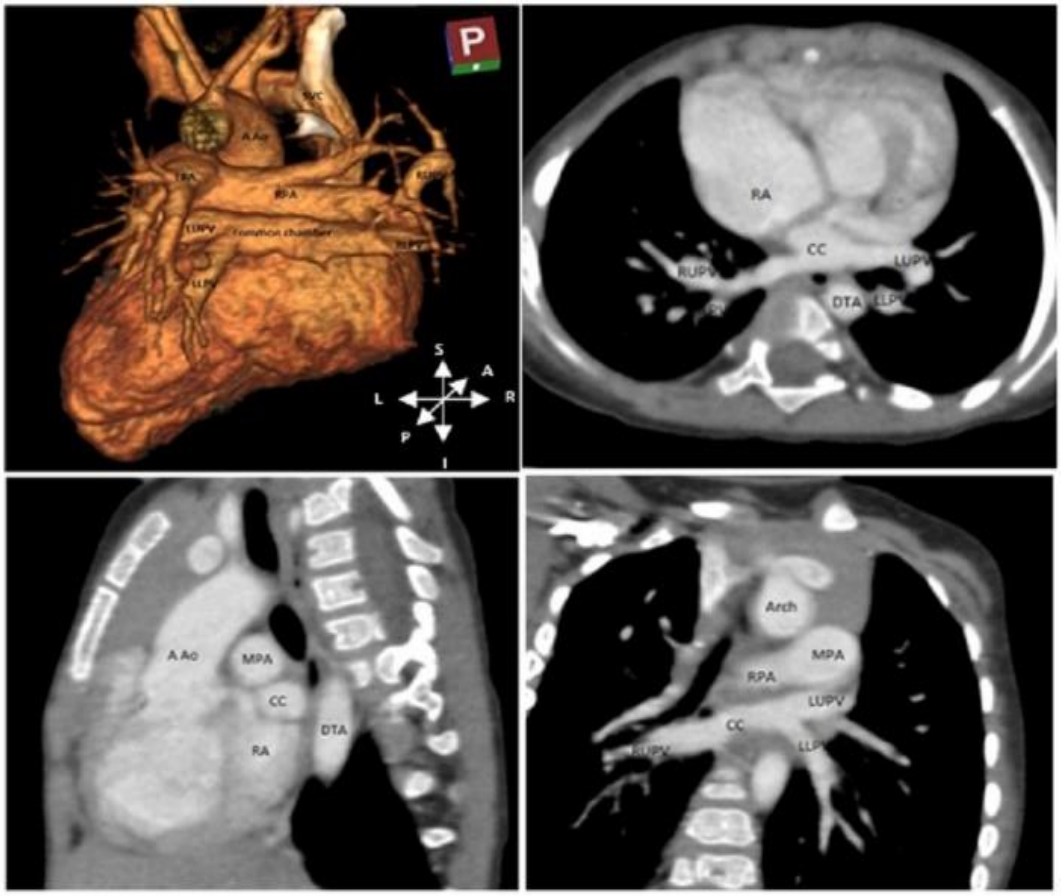

Fig. 3. In clock wise order from left upper corner, volume rendered computed tomography image, transverse, coronal and sagittal sections revealing formation of common chamber (CC) by all four pulmonary veins behind small left atrium (LA) and below the pulmonary artery branches. This chamber in turn drains 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 is frequently missed 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]. 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 [5,6]. Supracardiac and cardiac type TAPVC are more common than infracardiac.

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 importance to 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 because reduced 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 [7]. 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.

4. CONCLUSION

Being a rare congenital cardiac anomaly with clinical manifestations typical of isolated classical TOF, the diagnosis is frequently missed. First suspicion of TAPVC usually comes with certain clues in echocardiography including large RA coupled with small LA, presence of shelf like partition in LA, presence of separate chamber behind the LA, enlarged coronary sinus in absence of LSVC and demonstration of vertical vein. Successful treatment was achieved by anastomosing pulmonary venous confluence to the LA along with RVOTO and VSD repair.

CONSENT

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

ETHICAL APPROVAL

It is not applicable.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

REFERENCES

1. Talwar S, Choudhary SK, Shivaprasad MB, Saxena A, Kothari SS, et al. Tetralogy of Fallot with total anomalous pulmonary venous drainage. Ann Thorac Surg. 2008; 86:1937–40.
2. Ahmad Y, Mossa H. A 20-year follow-up of successful surgical management for a complex case of pentalogy of Fallot and dextrocardia with systemic and pulmonary venous anomalies: A rare case report. Int J Surg Case Rep. 2023;110: 108672.
DOI: 10.1016/j.ijscr.2023.108672.
Epub 2023 Aug 16.
PMID: 37598487; PMCID: PMC10469523.
3. Jeewa A, Mann GS, Hosking MC. Tetralogy of Fallot with absent pulmonary valve and obstructed totally anomalous pulmonary venous connection. Cardiol Young. 2007;17:551–3.
4. Festa P, Lamia AA, Murzi B, Bini MR. Tetralogy of fallot with left heart hypoplasia, total anomalous pulmonary venous return, and right lung hypoplasia: Role of magnetic resonance imaging. Pediatr Cardiol. 2005; 26:467–9.
5. Buckingham M, Meilhac S, Zaffran S. Building the mammalian heart from two sources of myocardial cells. Nat Rev Genet. 2005;6:826-35.
6. 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.
7. Maekawa Y, Miyahara Y, Yoshizumi K, Kawada M, Minami T, Sato T, Yokomizo A, Oka K, Furui S, Kataoka K. Tetralogy of Fallot with total anomalous pulmonary vein return and atrial septal defect: Successful two-staged surgical management; report of a case. Kyobu Geka. 2016;69(10):858-61. Japanese.
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