

PULMONARY ATRESIA WITH VENTRICULAR SEPTAL DEFECT (PA-VSD) IN ADOLESCENCE; AN EXTREME FORM OF CLASSIC TETRALOGY OF FALLOT

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ABSTRACT

Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease in all age groups, constituting approximately 8% of the congenital heart disease overall. Classic TOF consists of ventricular septal defect, right ventricular outflow tract obstruction, right ventricular hypertrophy, and overriding aorta. In contrast, PA-VSD consists of atresia of right ventricular outflow tract along with remaining three features of classic tetralogy of Fallot. It is both clinically and radiologically distinct from regular TOF. It comprises 5–10% of all tetralogy complexes. Survival rates in TOF with pulmonary atresia without surgical repair reported in the literature are as low as 50% at 1 year of age and 8% at 10 years. We present a rare case of uncorrected TOF with pulmonary atresia in adolescent female and describing the contrast-enhanced CT and angiographic features of this entity.

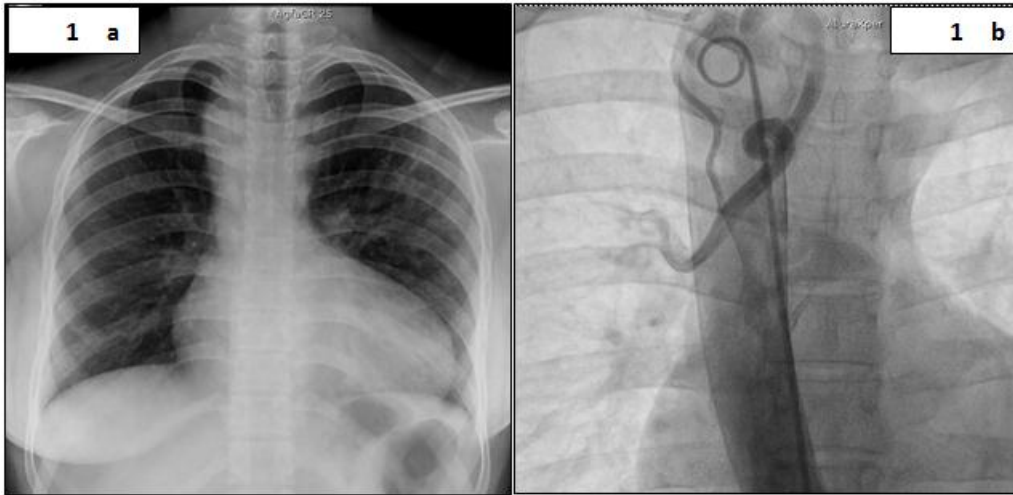
KEYWORDS: Congenital heart malformation, tetralogy of Fallot, CT angiography, MAPCAs.

INTRODUCTION

Case Report: We report a case of a 16 year-old female patient with uncorrected TOF and pulmonary atresia presenting to our cardiology department with class II (NYHA classification) shortness of breath. There was no history of haemoptysis/syncope/transient ischemic attack. On physical examination there was no pallor, cyanosis, clubbing, icterus or pedal edema. Her pulse rate was 72 and blood pressure was 110/60. On echocardiographic evaluation there was TOF morphology with RVOT atresia; pulmonary arteries were hypoplastic with maintained blood flow through MAPCAs.

On admission, a contrast-enhanced computed tomography (CT) of the chest was performed. The CT study shows that RVOT is atretic with maintained flow in hypoplastic main pulmonary artery through major aortopulmonary collateral arteries (MAPCAs) which are arising from aortic arch and descending aorta and ectatic left subclavian artery, a perimembranous ventricular septal defect (VSD) of 2.1 cm diameter, and right

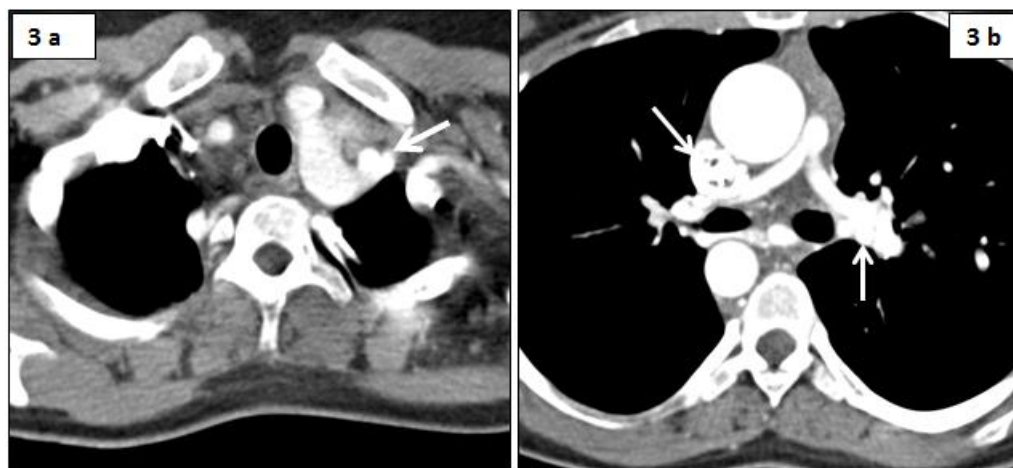
ventricular hypertrophy (RVH). There was no atrial septal defect/patent ductus arteriosus. Right-sided aortic arch with mirror image branching pattern is seen. Overriding of the aorta is noted (50%). Right coronary artery is arising from left anterior descending artery. Proximal left coronary artery is dilated and tortuous.



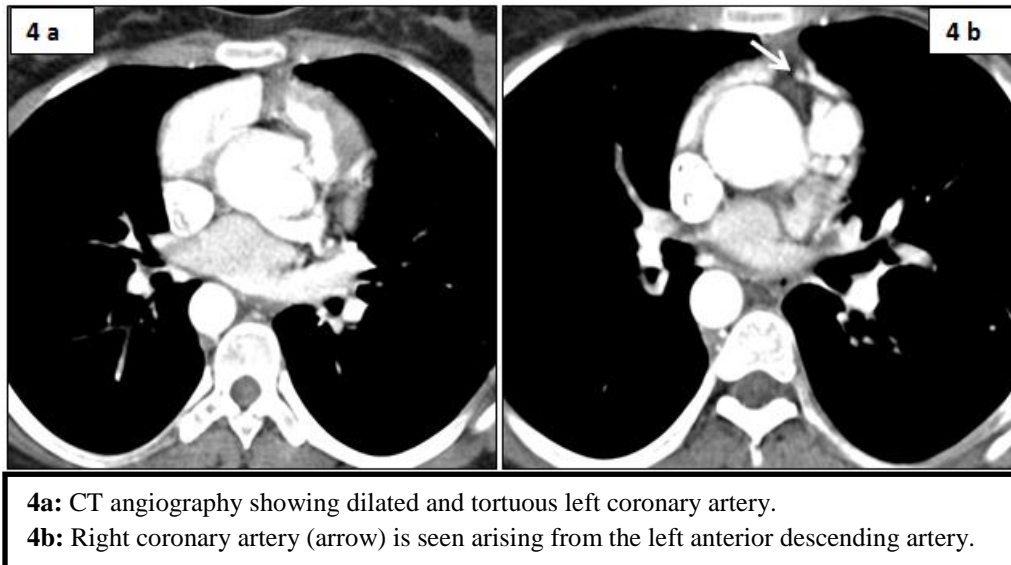
1a: Frontal chest x-ray showing paucity of hilar vessels and vascular markings in both the lung fields. Pulmonary bay is empty. There is also right ventricular hypertrophy.
1b: Angiogram showing a MAPCA arising from the aortic arch and supplying right pulmonary artery.



2 a: CT angiography showing right side aortic arch and MAPCAs (Arrow).
2 b: Overriding of aorta and perimembranous ventricular septal defect. Dilated right atrium and right ventricle hypertrophy is also seen.



3a: CT angiography showing mirror image branching pattern and ectatic left subclavian artery giving origin to MAPCAs (arrow).
3b: RVOT is atretic with hypoplastic main, right and left pulmonary artery with maintained flow through MAPCAs (arrows).



DISCUSSION

TOF is the most common cyanotic congenital heart disease overall in all age groups, comprising approximately 8% of congenital heart disease overall. It is the result of a single developmental abnormality, the misalignment of the crista supraventricularis with associated underdevelopment of the infundibulum. This abnormality results in ventricular septal defect, anterior displacement of the aortic valve, right ventricular outflow tract obstruction, and consequent right ventricular hypertrophy.^[1,2] A vast spectrum of pulmonary artery abnormalities can be seen in patients with tetralogy of Fallot. In mild cases, there is a ventricular septal defect and mild pulmonary valve stenosis, which is known as “pink” tetralogy, which may be asymptomatic. On other end of the spectrum there is complete pulmonary artery atresia with absence of the main pulmonary arteries, also known as pseudotruncus arteriosus. There are several degrees of severity in between these two extremes of the spectrum. Congenital Heart Surgeons Society proposed a practical classification based on complexity of pulmonary blood supply which in turn indicates the complexity of surgical repair.^[3]

Type A: Native PAs present, pulmonary vascular supply through PDA and no APCs.

Type B: Native PAs and APCs present.

Type C: No native PAs, pulmonary blood supply through APCs only.

Other commonly associated anomalies are right-sided aortic arch, which is present in 25% of cases, and atrial septal defect, which is present in approximately 5% of cases. Ventricular septal defect, persistent left superior vena cava, coronary artery abnormalities, and aberrant right subclavian artery also have been noted. In patients with this condition, systemic-pulmonary collateral vessels and a right-to-left shunt defect are essential for survival. Classic TOF consists of an interventricular communication, a biventricular connection of the aortic

root, which overrides the muscular ventricular septum an obstruction of the right ventricular outflow tract, and RVH.^[4] The variants of TOF include TOF with a patent foramen ovale/ASD (pentalogy), TOF with absent pulmonary valve, and TOF with pulmonary atresia. TOF with pulmonary atresia is clinically and radiologically distinct from regular TOF. It comprises 5–10% of all tetralogy complexes.^[5] The anatomy of central pulmonary arteries is often abnormal. MAPCA are systemic-to-pulmonary collateral arteries representing remnants of the embryonic ventral splanchnic arteries and provide an alternative pulmonary blood supply in patients with TOF and pulmonary atresia. These embryonic vessels normally regress concomitantly with the formation of the normal pulmonary arterial system in the first weeks of gestation, whereas in patients with pulmonary atresia they persist.^[6] Survival rates in TOF with pulmonary atresia without surgical repair reported in the literature are as low as 50% at 1 year of age and 8% at 10 years.^[7] Our case shows an adolescent survivor of uncorrected TOF with pulmonary atresia, the oldest survivor ever reported in the literature being 59 years old.^[4] As postulated by Fukui *et al.* the survival might be primarily dependent on the adequacy of pulmonary blood flow derived from MAPCAs.^[4]

CONCLUSION

TOF with pulmonary atresia is a rare entity of congenital heart disease presenting typical findings on cross-sectional imaging including VSD, right ventricular outflow tract obstruction, right ventricular hypertrophy, overriding aorta, and MAPCAs.

REFERENCES

1. Mirowitz SA, Gutierrez FR, Canter CE, Vannier, MW. Tetralogy of Fallot: MR findings. *Radiology*, 1989; 171(1): 207–212.
2. Van Praagh R, Van Praagh S, Nebesar RA, Muster AJ, Sinha SN, Paul MH. Tetralogy of Fallot:

- underdevelopment of the pulmonary infundibulum and its sequelae. *Am J Cardiol*, 1970; 26: 25–33.
3. Tchervenkov CI, Roy N. Congenital heart disease nomenclature and database project: Pulmonary atresia - ventricular septal defect. *Ann Thoracic Surg*, 2000; 69: S97-S105.
 4. Becker AE, Connor M, Anderson RH. Tetralogy of Fallot: a morphometric and geometric study. *Am J Cardiol*, 1975; 35: 402–412.
 5. Fukui D, Kai H, Takeuchi T, et al. Longest survivor of pulmonary atresia with ventricular septal defect: well developed major aortopulmonary collateral arteries demonstrated by multidetector computed tomography. *Circulation*, 2011; 124: 2155–2157.
 6. Boshoff D, Gewillig M. A review of the options for treatment of major aortopulmonary collateral arteries in the setting of tetralogy of Fallot with pulmonary atresia. *Cardiol Young*, 2006; 16: 212–220.
 7. Bertranou EG, Blackstone EH, Hazelrig JB, et al. Life expectancy without surgery in tetralogy of Fallot. *Am J Cardiol*, 1978; 42: 458–466.