IMAGES **in PAEDIATRIC CARDIOLOGY**

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Congenital absence of pulmonary valve syndrome (APV) represents a fascinating and unique variant of congenital heart disease. It was Chever in 1847 who first described this unique structural heart defect. The anatomic features consist of an incompletely formed, rudimentary pulmonary valve that is both stenotic and regurgitant, massively dilated pulmonary arteries and a large malaligned outlet ventricular septal defect. There is an association of this defect with Tetralogy of Fallot (ToF) due to which this condition is often referred to as Tetralogy of Fallot/absent pulmonary valve syndrome. Another characteristic feature is that there is virtually always absence of a patent ductus arteriosus. That has been hypothesized as being responsible for the pathogenesis of pulmonary artery dysplasia.[1]

APV syndrome is an extremely rare entity. It occurs in only 2.5% of patients diagnosed with ToF and in a 75% of these patients 22q11 deletion is identified.[1]

Diagnosis by echocardiography demonstrates a stenotic and malformed pulmonary annulus with remnants of a nonfunctioning, rudimentary valve. A large malaligned VSD is present. The most striking finding is the massively dilated main pulmonary artery and proximal pulmonary artery branches. Color Doppler will demonstrate systolic aliasing in the main pulmonary artery and diastolic aliasing in the right ventricular outflow truck as a result of pulmonary stenosis and regurgitation. It is unlikely that a patent ductus arteriosus is present and a thorough examination for major aortopulmonary collaterals should be performed.

The similarities of this congenital heart defect with tetralogy of Fallot makes the diagnosis challenging with the striking difference of the massively dilated pulmonary artery branches instead of the hypoplastic ones that are present in Fallot.[2]

Fig 1. The echocardiographic examination demonstrates cardiac anatomy very similar to Tetralogy of Fallot.

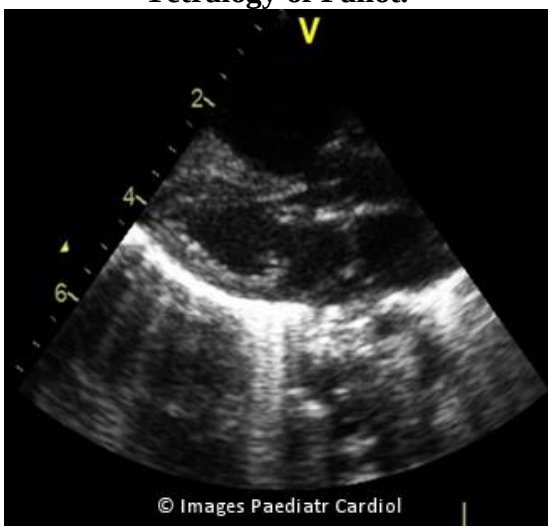


Fig. 2. The pulmonary valve is formed by dysplastic remnants of tissue and main and branch pulmonary arteries are massively dilated.

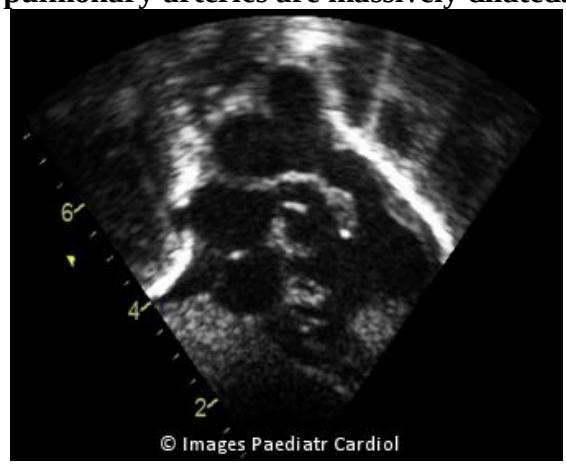
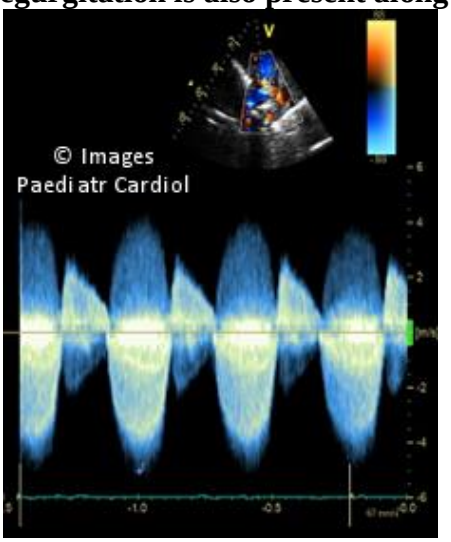


Fig 3. Severe pulmonary regurgitation is also present along with pulmonary stenosis.



References

1. Moss and Adams. Heart Disease in Infants, Children, and Adolescents. Philadelphia; Lippincott Williams and Wilkins, 2008.
2. 2.Eidem B, Cetta F, O' Leary PW. Echocardiography in Pediatric and Adult Congenital Heart Disease. Philadelphia; Lippincott Williams and Wilkins, 2010.

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