Article published online: 2021-07-19

Case Report

Fallot type of absent pulmonary valve syndrome - A case report

Divya B Desai, Neeti P Mathur, Arnab Marik

Department of Radiodiagnosis, D Y Patil Hospital and Research Center, Sector-5, Nerul, Navi Mumbai, Maharashtra, India

Correspondence: Dr. Divya B Desai, 204, Siddharth Apartment, Behind Kakdia Complex, Ghod Dod Road, Surat - 395 007, Gujarat, India.
E-mail: desaidivu2810@gmail.com

Abstract

Absent pulmonary valve syndrome (APVS) is a rare congenital cardiac malformation characterized by absent, dysplastic, or rudimentary pulmonary valve leaflets in association with other cardiac anomalies. It has an incidence of 3-6% in cases of tetralogy of Fallot (TOF) and 0.2-0.4% of live-born infants with congenital heart disease (CHD). Absent pulmonary valve leads to dilated main pulmonary artery; presenting as a pulsatile, paracardiac cystic lesion on antenatal ultrasound (USG). We report a case of this rare anomaly in association with ventricular septal defect (VSD), TOF, and left axis deviation of heart detected at 23 weeks of gestation.

Key words: APVS; IUGR; pulmonary valve; TOF; VSD

Introduction

Absent pulmonary valve syndrome (APVS) is a complex congenital cardiac syndrome first described by Cheevers in 1847.[1] It comprises of absent, dysplastic, or rudimentary pulmonary valvular leaflets with resultant regurgitation leading to dilatation of the main pulmonary arteries (MPAs) and branch pulmonary arteries (PAs).[2-4]

APVS is typically considered as a subgroup of tetralogy of Fallot (TOF). It is classified on the basis of presence or absence of ventricular septal defect (VSD) and features of TOF into Fallot type and non-Fallot type of APVS.[5,6] The incidence of APVS is rare as it accounts for 3-6% of cases of TOF and 0.2-0.4% of live-born infants with CHD.[5,7]

Access this article online	
Quick Response Code:	Website: www.ijri.org
	DOI: 10.4103/ijri.IJRI_508_19

Case Report

A 23-year-old primigravida presented for scheduled prenatal USG scan. USG revealed a single fetus of about 23 weeks' gestation.

Findings

On grayscale

- The grayscale scan revealed abdominal situs solitus (liver on the right side and fundic bubble on the left side) with left cardiac axis deviation. The fetus also had a pulsatile para cardiac cystic lesion that prompted us to perform dedicated fetal echocardiography.
- The thymus appeared mildly hypoplastic with fetal IUGR and oligohydramnios.

Cite this article as: Desai DB, Mathur NP, Marik A. Fallot type of absent pulmonary valve syndrome - A case report. Indian J Radiol Imaging 2020;30:240-3.

Received: 20-Dec-2019 **Revised:** 13-Feb-2020
Accepted: 19-Feb-2020 **Published:** 13-Jul-2020

- On fetal echocardiography, the following observations were made:
  - Apical four-chamber view appeared abnormal with dilated right ventricle (RV). The RV was seen to open into the pulsatile cystic lesion, which was thus confirmed to be the dilated MPA along with the right and left PAs. Assessment of the pulmonary annulus failed to reveal pulmonary valve (PV) echoes. Thus, the RV dilatation was attributed to volume overload from the insufficient PV. The ductus arteriosus was also not visualized despite an extensive targeted scan for its demonstration. An echogenic focus was noted in the left ventricle (LV) [Figure 1]
  - The five-chamber view revealed VSD with overriding of aorta and non-dilated aortic root [Figure 2]
  - Three vessel trachea view revealed massive dilatation of MPA, right and left PAs [Figure 3]
  - The aorta was seen arising from the LV
  - Sub-aortic VSD was seen
  - The superior and inferior vena cava drained normally into the right atrium. Atrioventricular concordance was noted with normal tricuspid and mitral valves.

On color and spectral doppler

- To-and-fro flow across the PV (a sign of stenosis and severe insufficiency) and the dilated PA was observed during systole and diastole of the RV [Figure 4]
- High velocities were recorded across PV annulus (PSV 190.4 cm/sec) [Figure 5]
- Aliasing was noted at the site of VSD on color Doppler study.

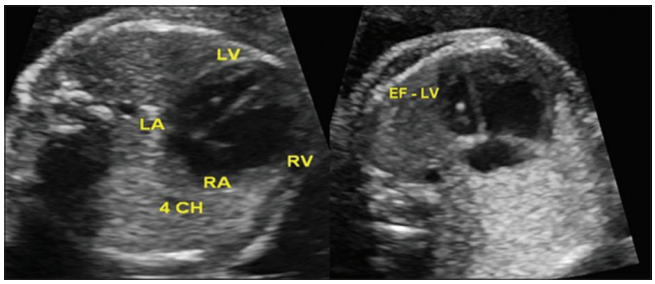

Figure 1: Apical-4 -chamber view: Dilated right ventricle [on right] and an echogenic focus (EF) in the left ventricle (LV) [on left]

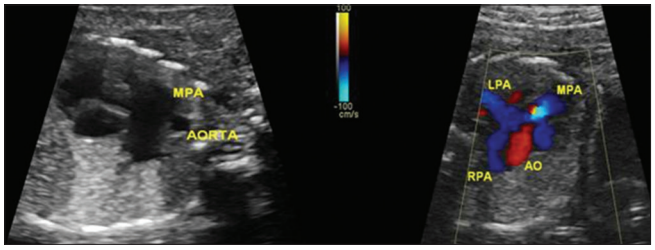

Figure 3: Three vessel trachea view: Massive dilatation of main pulmonary artery [on right] with non-dilated aorta and right and left pulmonary arteries [on left]

Hypoplasia of the left lung was noted with an approximately 2.5 × 2.5 cm hyperechoic lesion in the right hemi thorax receiving supply from the right PA. Other systems were unremarkable [Figure 6].

Based on the above imaging findings, a diagnosis of Fallot type of APVS was made. Karyotyping was offered for further evaluation but was refused by the patient. Patient counseling was done regarding the possibility of intrauterine (congestive heart failure and fetal hydrops) and neonatal complications (respiratory distress) and the need for multistage surgery. The patient opted for termination of pregnancy, however, autopsy of the fetus was not performed on patient request.

Discussion

Fallot type of APVS is the commonly seen type of APVS, while the non-Fallot type is the uncommon one.[5]

Despite having different morphology, they share common features, namely – rudimentary, dysplastic, or absent pulmonary valve; dilated MPA with or without dilatation of its branches; to-and-fro flow at the site of the absent

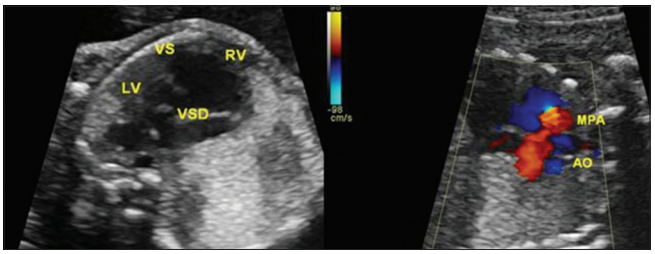

Figure 2: Five chamber view: Ventricular septal defect (VSD) [on right] with overriding of aorta [on left]

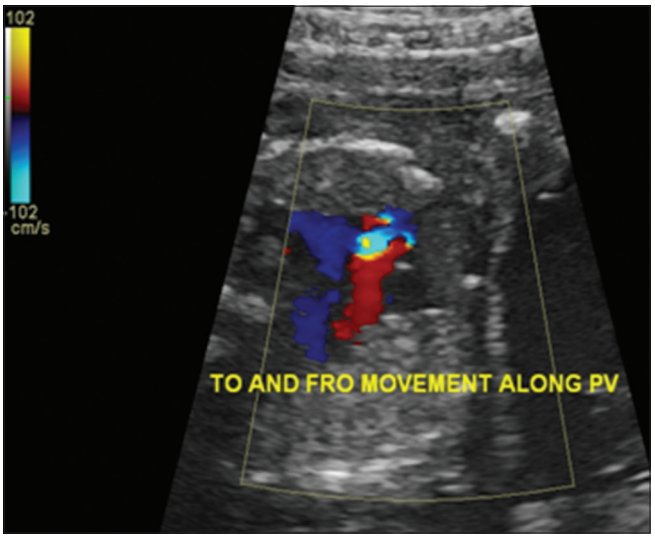

Figure 4: Color Doppler: To and fro movement along the pulmonary valve (PV) as a sign of stenosis and severe insufficiency

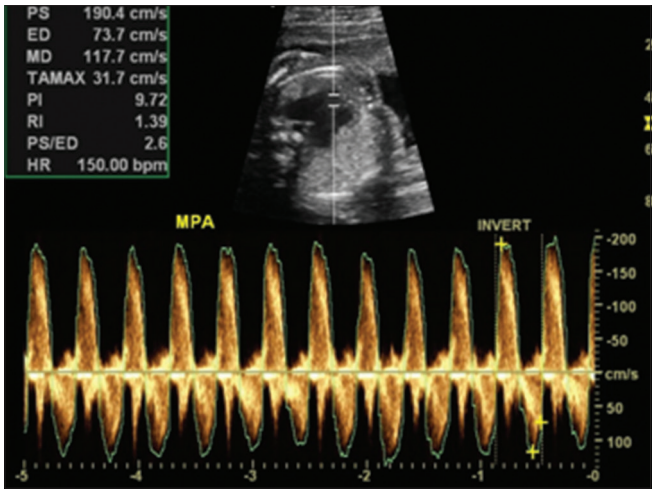

Figure 5: Spectral Doppler: High velocities across pulmonary valve annulus. Velocity 190.4 cm/sec

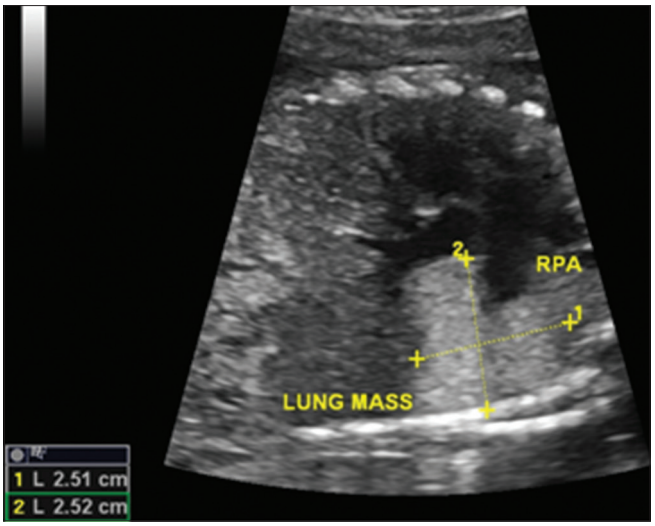

Figure 6: Hyperechoic lesion on the right lung region which is supplied by the right pulmonary artery

pulmonary valve and systolic pressure gradient across narrowed pulmonary valve.[5]

The Fallot type of APVS additionally has VSD, aortic overriding and ductal agenesis, which was first emphasized by Emmanoulides *et al*.[8]

As per few studies, most of the cases presenting with an intact ventricular septum commonly reveal a patent ductus arteriosus, with relatively small PAs and associated tricuspid atresia.[6,9] According to these studies, free communication between ventricles and aorta results in reduced atrial blood flow with increased ventricular end diastolic pressure, which may affect the cardiac function and development of the atrioventricular valve. Yeager *et al*. also suggest that in the presence of a VSD, these changes affect both ventricles, thereby resulting in a poor prognosis, as seen in our case.[6] In case of intact interventricular septum, these changes were restricted to the right ventricle.

The dilated PA morphology has been classified into two types[10]:

- Balloon type - bulbous expansion of the PA with indistinct bifurcation into bilateral PA
- Clover type - bulbous expansion of the PA with dilation of each of the bifurcated PA.

Kawazu *et al*. in their study found that the patients with balloon type of PA morphology were likely to require mechanical ventilation and had a poorer prognosis than patients with clover type of morphology.[10]

A grossly dilated PA can cause compression of the tracheobronchial tree and the esophagus, obstructing normal amniotic fluid circulation resulting in polyhydramnios, which is an indicator of poor prognosis.[11] Our patient did not show any polyhydramnios.

Volpe *et al*. found that neonatal bronchomalacia was a significant indicator of poor postnatal prognosis with cardiomegaly and marked branch pulmonary dilatation in a fetus with APVS indicating a high probability of occurrence of bronchomalacia.[12]

Fallot type of APVS has been associated with 22q11.2 microdeletion.[13-15] It has been found that presence of thymic abnormality (hypoplasia or aplasia) combined with intrauterine growth restriction (IUGR) associated with additional aortic arch anomalies had greater than 90% sensitivity and 100% specificity for this microdeletion.[15] Our fetus had thymic hypoplasia and features of IUGR without additional aortic arch abnormalities and hence was suspected to have 22q11.2 microdeletion.

Hypoplasia of the left lung with a hyperechoic lesion on the right lung region, which is seen supplied by the right PA was also noted. We suspected it to be either a sequestered lung or secondary to bronchial atresia. As autopsy was refused by the patient, a conclusive diagnosis of pulmonary abnormality was not achieved. Magnetic resonance imaging (MRI) was also suggested for confirmation of these findings, however, this non-invasive modality could not be availed due to the poor financial status of the patient.

In our patient, the fetus had a grave prognosis as ductal agenesis and a large VSD were present resulting in pressure changes in both ventricles with severe pulmonary arterial dilatation and early cardiac failure. The diagnosis was obtained on antenatal USG and echocardiography at the relatively early stage of 23 weeks of gestation. These findings were consistent with the findings reported in previous studies describing Fallot type of APVS.

Although few studies have revealed positive outcome after surgery,[2,16] Fallot type of APVS generally has a poorer outcome.[12,17,18]

Conclusions

A paracardiac cystic, pulsatile lesion in the fetus should alert the sonologist to the possibility of APVS. Classification into Fallot and non-Fallot type of APVS is important as the prognosis of former is poorer than latter. Presence of accessory findings like polyhydramnios and pulmonary hypoplasia indicate a poorer outcome. Soft markers like thymic abnormality (hypoplasia or aplasia) and/or IUGR associated with additional aortic arch anomalies need to be searched for in conotruncal anomalies to rule out 22q11.2 chromosomal microdeletion.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Cheevers N. Raaetraaecissement congenital de l" orifice pulmonaire. Arch Med Fourth Ser 1847;15:488.
2. Godart F, Houyel L, Lacour-Gayet F, Serraf A, Sousa-Uva M, Bruniaux J, *et al*. Absent pulmonary valve syndrome: Surgical treatment and considerations. Ann Thorac Surg [Internet] 1996;62:136-42. Available from: https://linkinghub.elsevier.com/retrieve/p ii/0003497596002767. [Last cited on 2019 Jul 28].
3. Bhupali AN, Patankar KB, Prasad S, Patil JK, Tamhane A. Absent pulmonary valve syndrome with tetralogy of Fallot detected at an early gestational age of 27 weeks-A case report. Indian Heart J 2013;65:191-3.
4. Pinsky WW, Nihill MR, Mullins CE, Harrison G, McNamara DG. The absent pulmonary valve syndrome. Considerations of management. Circulation 1978;57:159-62.
5. Grewal DS, Chamoli SC, Saxena S. Absent pulmonary valve syndrome-Antenatal diagnosis. Med journal, Armed Forces India 2014;70:198-200.
6. Yeager SB, Van Der Velde ME, Waters BL, Sanders SP. Prenatal role of the ductus arteriosus in absent pulmonary valve syndrome. Echocardiography 2002;19:489-93.
7. Yaman C, Arzt W, Tulzer G, Tews G. [Tetralogy of Fallot with absent pulmonary valve--prenatal diagnosis and management in the 2nd trimester]. Geburtshilfe Frauenheilkd 1996;56:563-5.
8. Emmanouilides GC, Thanopoulos B, Siassi B, Fishbein M. "Agenesis" of ductus arteriosus associated with the syndrome of tetralogy of fallot and absent pulmonary valve. Am J Cardiol 1976;37:403-9.
9. Joshi AN, Rane HS, Kamble RC, Mestry PJ, Maniar H, Shah Y. Prenatal diagnosis of absent pulmonary valve syndrome. J Ultrasound Med 2010;29:823-9.
10. Kawazu Y, Inamura N, Ishii R, Terashima Y, Hamamichi Y, Kayatani F, *et al*. Prognosis in tetralogy of Fallot with absent pulmonary valve. Pediatr Int 2015;57:210-6.
11. Callan NA, Kan JS. Prenatal diagnosis of tetralogy of fallot with absent pulmonary valve. Am J Perinatol 1991;8:15-7.
12. Volpe P, Paladini D, Marasini M, Buonadonna AL, Russo MG, Caruso G, *et al*. Characteristics, associations and outcome of absent pulmonary valve syndrome in the fetus. Ultrasound Obstet Gynecol 2004;24:623-8.
13. Boudjemline Y, Fermont L, Le Bidois J, Lyonnet S, Sidi D, Bonnet D. Prevalence of 22q11 deletion in fetuses with conotruncal cardiac defects: A 6-year prospective study. J Pediatr 2001;138:520-4.
14. Chaoui R, Kalache KD, Heling KS, Tennstedt C, Bommer C, Körner H. Absent or hypoplastic thymus on ultrasound: Amarker for deletion 22q11.2 in fetal cardiac defects. Ultrasound Obstet Gynecol 2002;20:546-52.
15. Volpe P, Marasini M, Caruso G, Marzullo A, Buonadonna AL, Arciprete P, *et al*. 22q11 deletions in fetuses with malformations of the outflow tracts or interruption of the aortic arch: Impact of additional ultrasound signs. Prenat Diagn 2003;23:752-7.
16. Yong MS, Yim D, Brizard CP, Robertson T, Bullock A, D'Udekem Y, *et al*. Long-term outcomes of patients with absent pulmonary valve syndrome: 38 years of experience. Ann Thorac Surg 2014;97:1671-7.
17. Zucker N, Rozin I, Levitas A, Zalzstein E. Clinical presentation, natural history, and outcome of patients with the absent pulmonary valve syndrome.Cardiology in the l Young 2004;14:402-8. Cambridge University Press.
18. Razavi RS, Sharland GK, Simpson JM. Prenatal diagnosis by echocardiogram and outcome of absent pulmonary valve syndrome. Am J Cardiol 2003;91:429-32.