

# Prenatal ultrasonic diagnosis of absent pulmonary valve syndrome

## A case report

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### Abstract

**Rationale:** Absent pulmonary valve syndrome (APVS) is a rare congenital heart disease that is often associated with tetralogy of Fallot (TOF). Here, we report 2 cases of APVS associated with TOF diagnosed via fetal echocardiography and discuss their specific ultrasonographic characteristics.

**Patient concerns:** Two pregnant women with suspicion of fetal heart anomaly were referred from their local hospitals to our hospital for fetal malformation screening and detailed fetal echocardiography. Color and spectral Doppler flow imaging were utilized to evaluate the axis, size, situs, cardiac chambers, and both inflow and outflow tracts of the heart as well as the great arteries. Both cases had a severe dilatation of the pulmonary trunk and its branches and an absence or dysplasia of the pulmonary valve, which was associated with subaortic ventricular septal defect (VSD) with an overriding aorta. In addition, the fetus in case 1 showed a patent ductus arteriosus, and the fetus in case 2 showed arterial duct agenesis. Furthermore, color Doppler flow imaging showed a bi-directional multicolored flow signal in the pulmonary valve ring.

**Diagnoses:** Both fetuses were diagnosed with APVS associated with TOF.

**Interventions:** No therapeutic intervention was performed.

**Outcomes:** On the request of the pregnant women and their families, both fetuses were aborted.

**Lessons:** Although APVS is a rare congenital heart disease and often associated with TOF, it has an overall poor prognosis. Nowadays, it can be easily diagnosed via ultrasonography because of its typical ultrasonographic features, such as aneurysmal dilatation of pulmonary artery, massive regurgitation of the pulmonary valve, VSD, and an overriding aorta. Therefore, early fetal echocardiography screening should be performed for every fetus.

**Abbreviations:** APVS = absent pulmonary valve syndrome, TOF = tetralogy of Fallot, VSD = ventricular septal defect.

**Keywords:** absent pulmonary valve syndrome, arterial duct agenesis, case report, prenatal diagnosis, tetralogy of Fallot

## 1. Introduction

Absent pulmonary valve syndrome (APVS) is a rare cardiac abnormality; its main characteristics are congenital dysplasia or

absence of a valve leaflet in the pulmonary area. APVS is also associated with expansion or aneurysmal dilatation of the main pulmonary artery and its branches resulting in valve stenosis and regurgitation.<sup>[1]</sup> Most of cases are complicated with the ventricular septal defect (VSD) and overriding aorta, so these cases are also known as the “absence of pulmonary valve associated with tetralogy of Fallot” (TOF).<sup>[2]</sup> The purpose of this case report is to describe the specific ultrasonographic features of APVS found in 2 fetuses.

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*Ethical approval: All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.*

*Informed consent: Informed consent was obtained from all individual participants included in the study.*

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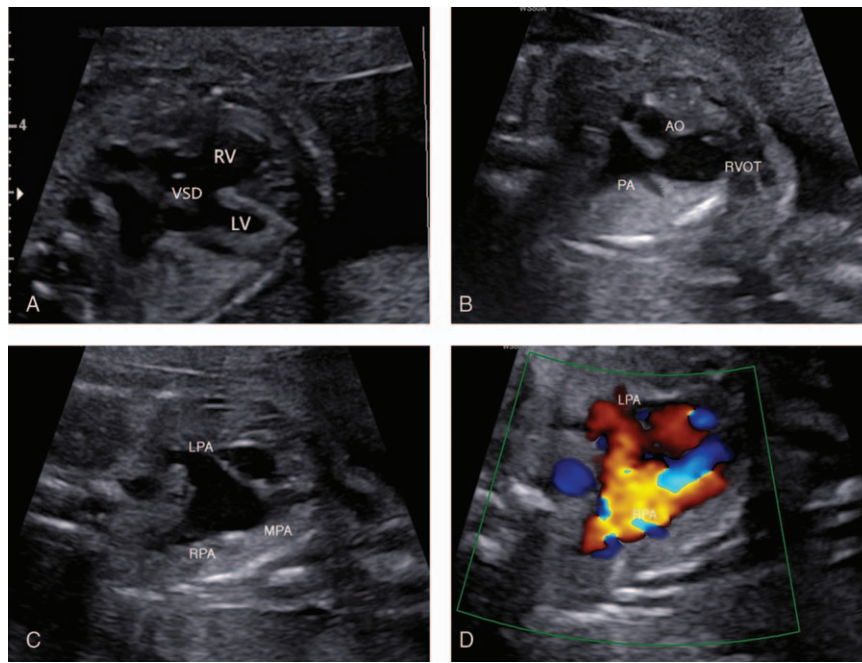
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## 2. Case presentation

### 2.1. General information

Two pregnant women who were in good health and had no malformations or genetic disorders in the family history were suspected of fetal heart anomaly in their local hospitals, so they were referred to our hospital for fetal malformation screening and fetal echocardiography. Case 1 was a 35-year-old woman, 2nd gravida at 23 weeks and 3 days; and case 2 was a 39-year-old woman, 3rd gravida at 22 weeks and 2 days. Fetal malformation screening found cardiac structural abnormality, but did not find any extracardiac abnormalities, in both fetuses. Then, detailed fetal echocardiography was performed with Samsung UGEOWS80A system with C2-6 transducer, and Philips EPIQ5 system with C5-1 transducer in OB/fetal heart mode.



**Figure 1.** Ultrasonographic features of APVS for case 1. Under the 4-chamber view, the bilateral atrioventricular valves are normal; the VSD and the overriding aorta are clear (A). Under the RVOT view, the pulmonary artery ring size is narrowed to about 3.5 mm (B), and the MPA is dilated with a diameter of about 11.2 mm (C). On color Doppler flow image, multicolored mosaic flow signal is seen in the MPA (D). AO=aorta, APVS=absent pulmonary valve syndrome, LPA=left pulmonary artery, LV=left ventricle, MPA=main pulmonary artery, PA=pulmonary artery, RPA=right pulmonary artery, RV=right ventricle, RVOT=right ventricular outflow tract, VSD=ventricular septal defect.

## 2.2. Ultrasonographic findings

In case 1, the apex cordis pointed to the lower left, and the 4 chambers, aortic arch, and superior and inferior vena cava were clearly viewed. A 4-chamber view (Fig. 1A) showed that the left and right atrioventricular valves were symmetrical, and the bilateral atrioventricular valves were normal. In addition, about a 5.5 mm VSD was seen at the upper part of the ventricular septum, and the aorta, whose diameter was about 4.6 mm, was overriding nearly 50% on the VSD. From the 3-vessel view and the right ventricular outflow tract view, the pulmonary artery ring size was narrowed to about 3.5 mm (Fig. 1B). Under the right ventricular outflow tract view (Fig. 1C), no obvious pulmonary valve activity was found, and the main pulmonary artery was dilated, with a diameter of about 11.2 mm and a length of about 12.6 mm. The left and right pulmonary arteries were clearly shown. Arterial duct could not be observed in all of the above sections. Color Doppler flow imaging showed the to-and-fro pattern of a multicolored mosaic flow signal that was seen in the main pulmonary artery within each heart cycle (Fig. 1D), with a flow rate of 2.9 m/s, a reverse directional blood flow, with a flow rate of about 2.2 m/s, from the main pulmonary artery to the outflow tract of the right ventricle that was seen during diastole, and a shunt signal shown at the ventricular level during systole.

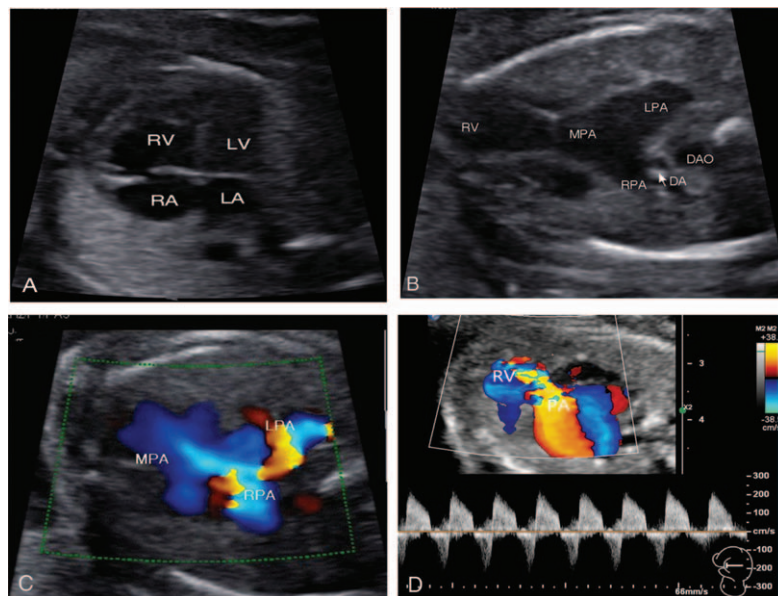
Case 2 presented with situs solitus, levocardia, and the normal atrioventricular connection. A 4-chamber view (Fig. 2A) showed that the left and right atrioventricular valves were symmetrical, the bilateral atrioventricular valves were noted, the VSD was about 4.7 mm, the aorta, whose dimension was about 3.9 mm, was overriding the VSD, and the pulmonary artery ring size was narrowed to about 2.4 mm. The 3-vessel view and the right ventricular outflow tract view found that the aorta overriding rate was about 50%. Under the right ventricular outflow tract view

(Fig. 2B), there was no obvious pulmonary valve activity, and the main pulmonary artery was dilated with a diameter of about 8.2 mm, the left and right pulmonary arteries were also significantly dilated with a diameter of about 7.3 and 7.2 mm, respectively, and the arterial duct was clearly shown. Color Doppler flow imaging showed the to-and-fro pattern of a multicolored mosaic flow signal that was seen in the main pulmonary artery within each heart cycle, with a flow rate of 2.7 m/s (Fig. 2C), a reverse directional blood flow, with a flow rate of about 2.1 m/s (Fig. 2D), from the main pulmonary artery to the outflow tract of the right ventricle that was seen during diastole, and a bi-directional shunt signal that was seen at the ventricular level during systole.

Considering all these findings above, the 2 cases were diagnosed with fetal complex congenital heart disease and APVS associated with TOF. Both pregnant women refused to undergo karyotype analysis and chose to terminate their pregnancies. Cardiac pathologic examination found no normal valve tissue in the pulmonary valve ring (Fig. 3), thus the ultrasonographic diagnosis was confirmed.

## 3. Discussion

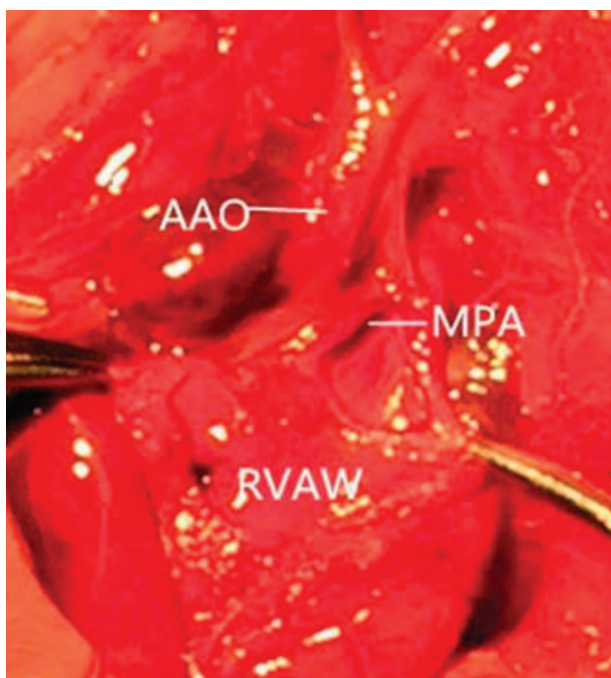
Simple APVS is rarely seen in clinical practice. The majority of APVS cases are often complicated with the VSD and overriding aorta, and among most of these cases, intrauterine closure of the arterial duct is shown by ultrasonography.<sup>[3]</sup> Therefore, these complicated APVS cases are also known as “absence of pulmonary valve associated with TOF.” The minority of APVS cases may also have several intracardiac malformations, such as tricuspid atresia,<sup>[4]</sup> right aortic arch,<sup>[5]</sup> and atrial septal defect. This congenital disease can nowadays be easily diagnosed by ultrasonography because of its specific ultrasonographic characteristics.



**Figure 2.** Ultrasonographic features of APVS for case 2. Under the 4-chamber view, the left and right atrioventricular valves are symmetrical (A). Under RVOT view, no obvious pulmonary valve activity is seen, the diameter of PA is about 8.2 mm, and the arterial duct is clearly shown (B). On color Doppler flow image, multicolored mosaic flow signal is seen in the MPA (C). On spectral Doppler flow image, antegrade and retrograde blood flow in the pulmonary trunk, and the right ventricle are shown (D). APVS=absent pulmonary valve syndrome, DA=arterial duct, DAO=descending aorta, LA=left atrium, LPA=left pulmonary artery, LV=left ventricle, MPA=mean pulmonary artery, PA=pulmonary artery, RA=right atrium, RPA=right pulmonary artery, RV=right ventricle, RVOT=right ventricular outflow tract, VSD=ventricular septal defect.

(1) A 3-vessel view and the right ventricular outflow tract view show a severe dilatation of the pulmonary trunk and its branches, with absence or dysplasia of the pulmonary valve.

(2) CDPI shows a bi-directional multicolored flow signal at the pulmonary valve ring.  
 (3) A 4-chamber view shows the enlargement of heart, especially the enlargement of the right ventricle.



**Figure 3.** Aneurysmal dilatation of the main pulmonary artery without normal pulmonary valve tissue at pulmonary valve ring. AAO=ascending aorta, MPA=mean pulmonary artery, RVAW=right ventricular anterior wall.

Therefore, if aneurysmal dilatation of the pulmonary artery and massive regurgitation of the pulmonary valve are observed, APVS should be taken into consideration, and then other structures of the fetal heart need to be further observed, such as ventricular septum, tricuspid valve, and arterial duct, to rule out other concomitant malformation. Furthermore, APVS is also often associated with defects in extracardiac organs, such as the polycystic kidneys, lip cleft, and palate cleft.<sup>[6]</sup> No extracardiac abnormality was found in either of our 2 cases. APVS can be differentiated from pulmonary artery stenosis or insufficiency by its absence of pulmonary valves and aneurysmal dilatation of the pulmonary trunk and its branches.<sup>[7]</sup>

The ultrasonographic characteristics in our 2 cases were very similar and both showed APVS associated with TOF. However, the arterial duct was found in case 2, but not in case 1. For cases of APVS with TOF, although the arterial duct occurs very rarely, as in case 1, the existence of the ductus arteriosus increases the flow in the aorta and the main pulmonary artery.<sup>[8]</sup> Becker et al<sup>[3]</sup> considered that the fetal arterial duct may close between 14 and 21 weeks of pregnancy in some cases, so arterial ducts usually cannot be seen by ultrasonography in the fetuses of middle-late pregnancies with APVS. However, some scholars found the presence of the arterial duct by ultrasonography in a small subset of APVS cases of late pregnancy, and thus thought that the degree of pulmonary artery dilatation was related to the presence or absence of the arterial duct.<sup>[9]</sup> Nevertheless, whether the closure of the arterial duct causes dysplasia or absence of the pulmonary valve or vice versa, or there is no hemodynamic relation between them is still not clearly defined.<sup>[3,8,9]</sup>



Some cases of APVS associated with TOF have microdeletion of 22q11.2.<sup>[1,5,9]</sup> However, both our cases refused to participate in karyotype analysis and no karyotype analysis was performed for 2 fetuses either. Hence, we do not know whether the 2 fetuses also had microdeletion of 22q11.2.

APVS has an overall poor prognosis, and there is no effective intrauterine treatment. According to the study by Razavi et al,<sup>[8]</sup> among 20 cases of APVS, 6 neonates died even after surgery, and only 3 survived more than 1 year. Furthermore, the prognosis is not significantly affected by whether the disease is diagnosed prenatally or postnatally.<sup>[11]</sup> However, APVS cases with different complications have different outcomes.<sup>[8,9]</sup> In addition, fetus with an isolated APVS or an intact ventricular septum has a relatively better prognosis.<sup>[10]</sup>

#### 4. Conclusions

APVS is a rare congenital heart disease and often associated with TOF, but has an overall poor prognosis. Thanks to its unique features, such as aneurysmal dilatation of the pulmonary artery, massive regurgitation of the pulmonary valve, VSD, and an overriding aorta, APVS can be easily diagnosed by ultrasonography. Prenatal screening by ultrasonography not only detects APVS, but also shows the relationship between APVS and its concomitant complications, if they exist, thereby helping estimate the possible outcomes of APVS, which is crucial for making clinical decisions. Therefore, early fetal malformation screening and echocardiography screening should be considered for each fetus.

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