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BRIEF COMMUNICATION

Cardinal Clues Conducive to the Diagnosis of Fetal Absent Pulmonary and Aortic Valves in the First Trimester: A Case Report

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Abstract Fetal absent pulmonary valve (APV) is a rare cardiac lesion. Prenatal reports of absent aortic valve (AAV) are even scarce. Further, a combination of this lesions have never been reported in the first trimester. The objective of our case report is to describe the prenatal ultrasound features of a case of APV co-existing with AAV. Following the detection of hydrops at 12^{+2} gestational weeks, a first trimester echocardiographic examination revealed situs solitus, sinus rhythm, normal septo-aortic continuity, presence of Ductus arteriosus, to- and fro-blood flow across both the pulmonary and the aortic orifices, 'a' wave reversal in the Ductus venosus, and reversed end diastolic flow in the umbilical artery; suggesting a diagnosis of APV with AAV. The couple opted for termination of the pregnancy. This case underscores the importance of meticulous utilization of color Doppler in the first trimester, especially in the background of increased nuchal translucency or hydrops.

Keywords Absent pulmonary valve · Absent aortic valve · Pulmonary stenosis · Pulmonary artery dilatation · Pulmonary regurgitation · Absent Ductus arteriosus

Introduction

Absent Pulmonary Valve (APV) is a rare cardiac lesion with an incidence of 0.2–0.4% of the postnatal [1], and 1% of the prenatal cohort of congenital heart diseases [2]. It is characterized by partial or complete agenesis of the pulmonary valve. The first case of APV was reported in 1867 [3]. Since then, consistent description in both postnatal and prenatal series has enhanced our understanding of its pathophysiology, extra cardiac and chromosomal associations [4]. Only few cases of Absent Aortic Valve (AAV) have been described prenatally, none in the first trimester [5]. The objective of our case report is to elucidate the ultrasound features of a case of APV co-existing with AAV in the first trimester. To the best of our knowledge, these combination of lesions have not been reported in the first trimester.

Case Report

A 22 years old Indian second gravida with a singleton pregnancy was found to have features of fetal hydrops at the 12^{+2} gestational week. Detailed fetal cardiac assessment demonstrated the following features (Fig. 1)—situs solitus, sinus rhythm with a heart rate of 140 beats per minute, and a cardiac axis of 23°. In the Three Vessel Trachea (3VT) view, to- and fro-motion of blood was noted across both the ductal and aortic arches. There was no obvious dilatation of the pulmonary arteries. The pulmonary Peak Systolic Velocity (PSV) was 30 cm/s. A reversal of the 'a' wave was noted in the ductus venosus. The umbilical artery Doppler showed reversed diastolic flow.

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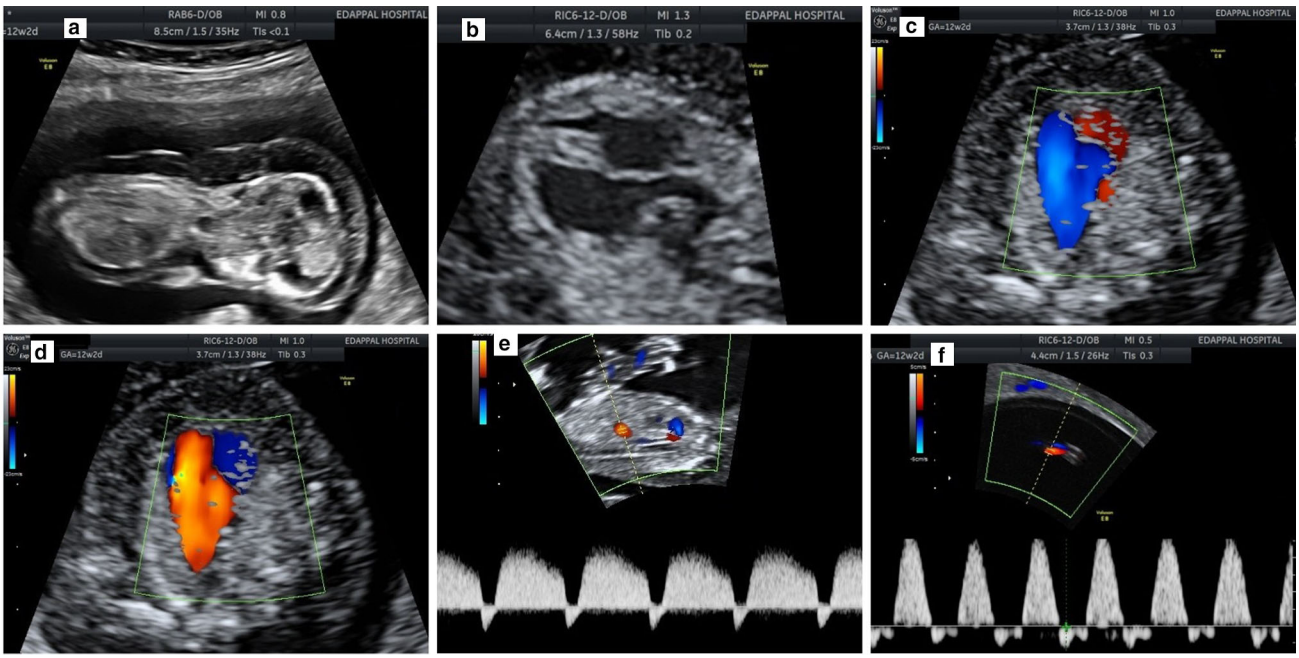

Fig. 1 First trimester ultrasound features of absent pulmonary and aortic valve. **a** Fetus at 12^{+2} weeks with edema all around the fetus (hydrops), **b** the left ventricular outflow view depicting septo-aortic continuity and ruling out TOF, **c** the 3VT view showing antegrade flow in both arches during systole, **d** retrograde flow in both arches during diastole due to absence of both the pulmonary and aortic valves, **e** reversal of 'a' wave in the ductus venosus Doppler, and **f** reversal of the end diastolic frequencies in the umbilical artery Doppler

Based on these findings, a diagnosis of APV/non-Tetralogy of Fallot (TOF) type, with patent Ductus Arteriosus (DA), and aortic valvular agenesis was made. The couple were informed of the findings, and a fetal karyotype was advised. The couple, however, opted for termination of the pregnancy.

Discussion

Embryologically, development of conotruncal cardiac anomalies including APV is linked to the abnormalities of the cephalic neural crest cells [6, 7]. Three anatomical variants of APV have been described—(1) APV with TOF (APV/TOF), (2) APV without TOF (APV/non-TOF), and (3) APV with Tricuspid Atresia (APV/TA) [8]. The APV/TOF is the most common type observed in over 80% cases, and is characterized by malalignment VSD, overriding aorta and absence of DA [9]. The APV/non-TOF has a normal DA with usually intact interventricular septum [1]. The APV/TA is extremely rare, and is characterized by tricuspid atresia or stenosis with right ventricular dysplasia [10]. The diagnosis of APV in the second trimester is based on aneurysmal dilatation of the main and branch pulmonary arteries, annular pulmonary stenosis, and pulmonary regurgitation across the unguarded pulmonary orifice manifesting as to- and fro-motion of blood [1].

However, the presentation of APV differs in the early gestation [4]. In the first trimester, APV/AAV usually presents as increased NT or hydrops [1]. The diagnosis of APV depends on the analysis of the spectral and color Doppler signs, as was depicted in our case. A to- and fro-motion of blood across the pulmonary orifice in color Doppler is pathognomonic of APV, however, is not trimester specific. This sign is explained by the antegrade flow across the pulmonary orifice during systole, and diastolic regurgitation across the unguarded orifice. An 'a' wave reversal is noted in the DV Doppler. Further, Reversed End Diastolic Velocities (REDV) are noted in the Umbilical Artery Doppler (UAD), which in 11–14 weeks is a characteristic sign of APV [11]. Usually, between 11 and 14 weeks, positive end diastolic flow appears in UAD [12]. The REDV in the first trimester is also noted in chromosomal anomalies, and in early fetal growth restriction [13, 14]. The to- and fro-motion of blood flow across the aorta, seen in our case, is explained by a co-existent absent aortic valve.

The APV/non-TOF type with patent DA has massive regurgitation resulting in bi-ventricular overload, and consequent hydrops. Majority of these cases do not reach second trimester. In APV/TOF type, since absence of DA limits regurgitant volume of blood into the ventricles, hydrops is observed less frequently [15]. Hydrops is also rare in APS/TA type due to atretic tricuspid valve [10].

The prognosis of APV depends on its type and associations. In general, the dilated pulmonary vessels can compress the bronchial tree resulting in bronchomalacia [1]. Hydrops is a common complication of APV/non-TOF type. The APV/TOF type is associated with 22q11 microdeletion in 25% cases [9]. A recent series has shown 86% survival in the isolated cases of APV [8].

In conclusion, early diagnosis of APV is associated with higher incidence of chromosomal abnormalities, hydrops and poorer prognosis. Hence, it is imperative for first trimester scans ($11–13^{+6}$ weeks) to have a color flow mapping of the four chambers and 3VT views, especially in the backdrop of hydrops/increased NT. Diligent inspection of Doppler signs can facilitate early cardiac diagnosis.

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