Case report

Absent pulmonary valve syndrome with tetralogy of Fallot detected at an early gestational age of 27 weeks – A case report

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ABSTRACT

Objective: Absent pulmonary valve syndrome (APVS) is a rare congenital anomaly, usually seen in association with a ventricular septal defect. It has been reported to occur in 3–6% of cases of tetralogy of Fallot (TOF). In this case report we discuss a case of absent pulmonary valve syndrome with tetralogy of Fallot that was detected in utero by fetal echocardiography at 27 weeks of gestation.

Case: A 20-year-old pregnant woman at 27 weeks of gestation referred to our Institute. She has no consanguineous history. We diagnosed the case as tetralogy of Fallot with absent pulmonary valves in fetal echocardiographic study.

Conclusion: We conclude that when a paracardiac cystic, pulsatile lesion with dilated pulmonary arteries are seen in the fetus in utero then other features associated with the syndrome, such as TOF and the presence or absence of the ductus arteriosus should be looked for. In our case there was no ductus arteriosus.

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1. Introduction

Absent pulmonary valve syndrome (APVS) is a rare congenital anomaly, usually seen in association with a ventricular septal defect. It has been reported to occur in 3–6% of cases of tetralogy of Fallot (TOF). Absence of the pulmonary valve results in a dilated main pulmonary artery, which can be seen as a cystic, pulsatile, paracardiac lesion on antenatal USG.
Such a lesion, though rare, can easily be detected. We report a case of this rare anomaly which was present in association with a TOF. The case was detected at 27 weeks of gestation. The prognosis depends on the respiratory complications.\textsuperscript{1} When APVS is associated with a ventricular septal defect, the physiologic and anatomic repercussions affect both ventricles, and cardiac performance can be critically impaired.\textsuperscript{2} We report a case of APVS that was detected by USG at 27 weeks of gestation; As of now the fetus is grossly normal and growth is corresponding to gestational age.

2. Case report

A 20-year-old primigravida presented for routine prenatal USG scanning. There was a single fetus of about 27 weeks gestation. The fetus showed a pulsatile cystic lesion located near the heart. We therefore performed a detailed fetal echocardiography, which revealed. The superior and inferior vena cava drained normally into the right atrium. Both Atria were normal. Atrioventricular concordance was noted. The tricuspid and mitral valves were normal. The right ventricle (RV) was seen to open into the pulsatile cystic lesion, which was thus confirmed to be the dilated main pulmonary artery along with the right and left pulmonary arteries. The pulmonary annulus showed no pulmonary valve (p-valve) echoes Fig. 1. There was back-and-forth flow across the RV and the diluted pulmonary artery during systole and diastole of the RV Fig. 2. The aorta was seen arising from the left ventricle (LV) and there was 50% overriding of aorta. A large subaortic ventricular septal defect (VSD) was seen Fig. 3. On Color Doppler study pulmonary regurgitation was noted Fig. 4. Other systems were unremarkable. Thus, a diagnosis of TOF with absent pulmonary valves was made.

3. Discussion

APVS is a complex syndrome comprising dysplasia/absence of pulmonary valvular leaflets, with resultant regurgitation and dilatation of the main and branch pulmonary arteries.\textsuperscript{3} The majority of these cases present with a VSD and features of TOF. The aneurysmal dilatation of the pulmonary artery often results in compression of the bronchial tree and esophagus, with consequent bronchomalacia and polyhydramnios. Volpe et al studied 21 fetuses with APVS for their associations and outcomes. Their study reveals an association of this syndrome with microdeletion of chromosome 22q11 in 25% of cases. They also suggest that bronchomalacia is commonly associated with cardiomegaly and dilatation of the pulmonary artery and results in poor prognosis.\textsuperscript{4} APVS, in the absence of VSD, is uncommon. As reported by Yeager et al, most of the cases presenting with an intact ventricular septum commonly reveal a patent ductus arteriosus, with relatively small pulmonary arteries and associated tricuspid atresia.\textsuperscript{2} According to their study, the free communication
between the ventricles and the aorta causes the blood flow to the atria to be reduced, while there is an increase in the ventricular end diastolic pressure; this may in turn affect the cardiac function and the development of the atrioventricular valve. Yeager et al also suggest that in the presence of a VSD these changes affect both the ventricles, thereby resulting in a poor prognosis, as seen in our case. A grossly dilated pulmonary artery can cause compression of the tracheobronchial tree and the esophagus. This obstructs the normal amniotic fluid circulation, causing polyhydramnios. As stated by Callan et al, the presence of polyhydramnios may indicate a poor prognosis.

4. Conclusion

We conclude that when a paracardiac cystic, pulsatile lesion is seen in the fetus in utero, APVS is an important differential diagnosis and other features associated with the syndrome, such as TOF and the presence or absence of the ductus arteriosus should be looked for.

REFERENCES