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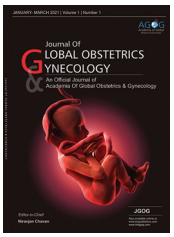
Case Report



Perinatal Diagnosis of the Tetralogy of Fallot with Absent Pulmonary Valve in a Pregnant Woman with Overt Diabetes of Mellitus – A Case Report

Alireza Golbabaei[1], Maasoumeh Saleh[2], Mahsa Naemi[2], Azadeh Rastegar[2], Mahboubeh Saleh[3]

[1]Department of Perinatology and Fetal Cardiology, Shariati Hospital, Tehran University of Medical Sciences, Tehran, Iran, [2]Department of Obstetrics and Gynecology, Shariati Hospital, Tehran University of Medical Sciences, Tehran, Iran, [3]Department of Obstetrics and Urogynecology, Fasa University of Medical Sciences, Fasa, Iran.

ABSTRACT

The tetralogy of Fallot (TOF) with absent pulmonary valve is a rare, congenital, cyanotic heart defect. Its characteristics are pulmonary stenosis and regurgitation, ventricular septal defect and aorta overriding, and huge dilation of pulmonary artery branches. We would like to report a case of the TOF and absent pulmonary valve in a 33-year-old G3-P1-L1 pregnant female at the gestational age of 18 weeks with a history of pre-pregnancy overt diabetes mellitus.

Key words: Absent pulmonary valve, Diabetes mellitus, Pregnancy, Tetralogy of Fallot

INTRODUCTION

Absent pulmonary valve syndrome is a rare congenital conotruncal anomaly.[1-6] Its main characteristics are congenital rudimentary dysplastic pulmonary valve or the absence of a valve leaflet in the pulmonary valve.[2,3]

It has been reported to occur in 3–6% of cases of tetralogy of Fallot (TOF), and it is considered as a variant of the TOF.[1,4,6] The absence of pulmonary valve results in a dilated main pulmonary artery and its branches (one or both), resulting in valve stenosis and regurgitation[1-5] which can be seen as a cystic, pulsatile, and pericardiac lesion.[1] Most of the cases are complicated with the malalignment type ventricular septal defect (VSD) and overriding aorta, so these cases are also known as the "absence of pulmonary valve associated to the TOF." On the other hand, its main association is through the TOF, microdeletion 22q11.2, and arterial duct agenesis[1,2,3,5] which have been associated to the significant perinatal mortality and morbidity as the dilated PA branches may result in bronchomalacia secondary to bronchial compression with substantial respiratory symptoms and high respiratory failure at birth in cases due to bronchial narrowing of severely dilated pulmonary arteries.[3,4,7]

In these patients, the prognosis depends on the respiratory complications.[6] After echocardiographic prenatal diagnosis, relatively high termination rates range from 30% to 43%, and 1-year mortality rates range from 67% to 75%. The perinatal risk factors for mortality include respiratory distress at birth, the presence of a genetic syndrome or abnormal karyotype, and the presence of hydrops fetalis. More recent series have reported better rates of survival, ranging from 72% to 86%. It may be due to better anticipatory planning at birth after prenatal diagnosis and improved surgical strategies for repair. As prenatal ultrasound screening improves, an interesting and relatively uncommon anomaly is being identified by increasing the frequency.[4]

CASE PRESENTATION

A 33-year-old G3-P1-L1 pregnant female referred to our clinic at her 18th gestational age. Her history was overt diabetes mellitus for 3 years ago. No additional risk factors have existed and her blood group was A positive. In fetal echocardiography, the orientation and position of heart were normal (levocardia and levoposition). Apical four-chamber view showed right ventricular hypertrophy [Figure 1] and enlargement. In the five-chamber view, large outlet type (subaortic) VSD and dilated aortic root (aorta overriding) were seen [Figure 2]. The short-axis view

Corresponding Author:
Maasoumeh Saleh, Department of Obstetrics and Gynecology, Shariati Hospital, Tehran University of Medical Sciences, Tehran, Iran.
E-mail: salehmaasoumeh@yahoo.com

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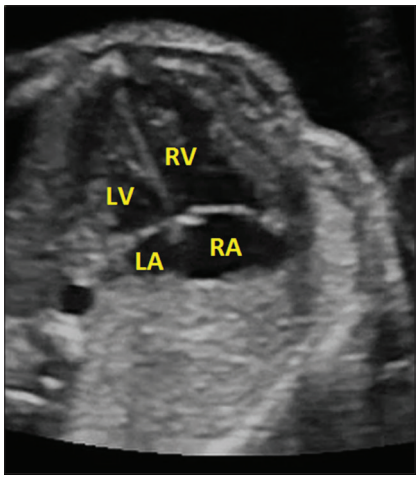

Figure 1: Apical four-chamber view shows the right ventricular hypertrophy and enlargement

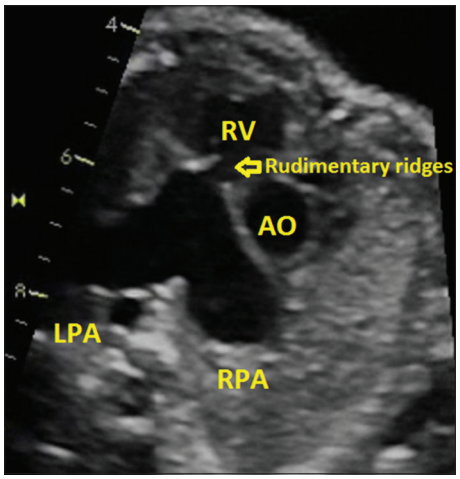

Figure 3: Short-axis view shows the massive dilation of pulmonary artery branches, rudimentary ridge (such as pulmonary leaflets)

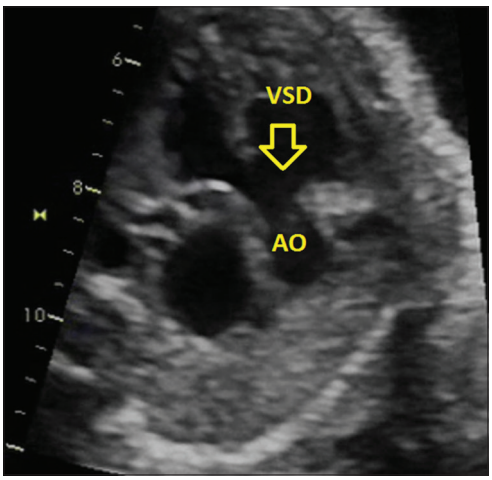

Figure 2: Five-chamber view shows the large outlet (subaortic ventricular septal defect) and aorta overriding

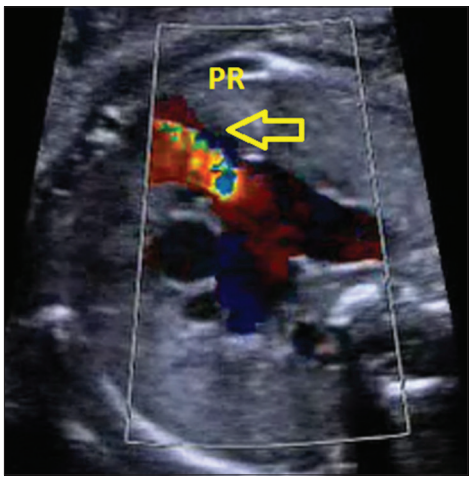

Figure 4: Color Doppler (short-axis view) shows the significant pulmonary valve regurgitation

showed a normal relationship between the aorta and pulmonary artery, but massive dilated pulmonary branches and rudimentary ridge (as pulmonary leaflets) were seen [Figure 3]. Furthermore, significant pulmonary valve regurgitation with color Doppler use was viewed [Figure 4]. When Doppler spectral positioned on pulmonary regurgitation direction, pulmonary stenosis and significant pulmonary regurgitation Doppler pattern were determined [Figure 5]. No other anomalies were observed. The termination of pregnancy was performed in the 19th weeks of the gestational age.

DISCUSSION

The absent pulmonary valve is a rare and complex syndrome comprising dysplasia/absence of pulmonary valvular leaflets, with resultant significant regurgitation (to and fro flow between the right ventricle and pulmonary valve), and marked dilatation in the main of pulmonary artery and branches.[1,2,6,7] PA dilatation occurs early in pregnancy.[1,3]

The majority of these cases present with a VSD and overriding of aorta. Therefore, these complicated cases are also known as "the absence of pulmonary valve associated to TOF (in more than 80%)."[1,2,3,7]

The minority of patients may also have several intracardiac malformations such as tricuspid atresia, right aortic arch, and atrial septal defect.[2,7] Our case was not associated to other intracardiac abnormalities.

The most unusual association of absent pulmonary valve occurs with tricuspid atresia/stenosis. A decade ago, Litovsky *et al*.[11] reported the postmortem findings of three children which reviewed the literature on 24 additional cases. In the first two fetal cases, only one was born and underwent staged single-ventricle surgery.[3]

This congenital disease can be easily diagnosed by ultrasound due to its specific ultrasonographic characteristics.[1] Conotruncal

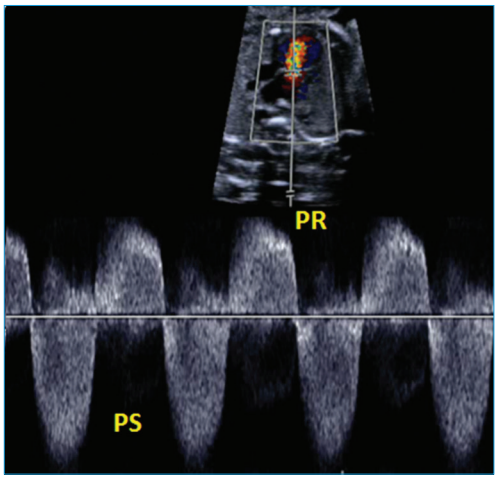

Figure 5: Doppler spectral in the pulmonary valve shows the pulmonary stenosis and pulmonary regurgitation Doppler pattern

anomalies can be diagnosed in prenatal life by fetal echocardiogram with a high degree of accuracy. An absent pulmonary valve can be suspected when we see any associated echocardiographic findings such as aneurysmal dilatation of pulmonary arteries and its branches appearing as "bow-tie"- or "balloon"-like hypoechoic shadow. Studies conducted by Andrew *et al.*[12] who showed that conotruncal anomalies pose a poor prognosis for the fetus, and 64% of the fetuses with a prenatal diagnosis of conotruncal anomalies died. The mortality is comparable with Allan *et al.*[13] study which showed mortality ranged from 50% to 65%.[5]

A 3-vessel view and short-axis view of great arteries show a severe dilatation of the pulmonary trunk and its branches, with the absence or dysplasia of pulmonary valve. Color Doppler shows a bidirectional flow at the pulmonary valve due to antegrade flow through pulmonary valve and valvular regurgitation. A 4-chamber view maybe shows the cardiomegaly, especially the enlargement of the right ventricle.[2] Cardiomegaly and significant pulmonary regurgitation were prominent in our patients.

The pulmonary artery's aneurysmal dilatation often results in compression of the bronchial tree and esophagus, with consequent respiratory failure and bronchomalacia.[1,3,6,7] In a study by Pinsky *et al.*,[14] 40% of children with absent pulmonary valve had developed respiratory distress during early infancy secondary to aneurysmal dilatation of pulmonary arteries compressing bronchi resulting in massive lobar emphysema. An autopsy done at neonatal death showed that ductus was almost always absent.[15] This obstructs the normal amniotic fluid circulation, causing polyhydramnios. Callan *et al.*[16] stated that polyhydramnios' presence might indicate a poor prognosis.[6] In our case, polyhydramnios was not seen. The severity of tracheobronchial tree compression is not predicted accurately by fetal echocardiography. Therefore, if aneurysmal dilatation of the pulmonary artery is observed, an absent pulmonary valve should be considered.[2,3] This anomaly is often associated to the defects in extracardiac organs such as the polycystic kidneys, lip cleft, and palate cleft.[2] No extracardiac abnormality was found in our case.

Volpe *et al.*[8] studied 21 fetuses with absent pulmonary valve for their associations and outcomes. They suggested that the pulmonary artery's dilatation is commonly associated to bronchomalacia and results in poor prognosis after birth.[1,6]

All cases of this anomaly carry a risk of fetal heart failure due to ventricular volume overload. Intrauterine demise preceded by fetal hydrops occurred in 7–15% of pregnancies. The most common genetic and chromosomal abnormality was a 22q11.2 deletion, emphasizing that chromosomal testing, including FISH analysis, should be offered, whenever this condition is detected.[2,3] However, our case refused to participate in karyotype analysis, and no karyotype analysis was performed.

Becker *et al.*[9] considered that the fetal ductus arteriosus may close between 14 and 21 weeks of pregnancy in some cases.[2] In our case, the ductus arteriosus was patent until pregnancy termination time.

The absent pulmonary valve has an overall poor prognosis, and there is no effective intrauterine treatment. Considering the study of Razavi *et al.*,[10] among 20 cases of the absent pulmonary valve, six neonates died even after surgery, and only three survived more than 1 year. Furthermore, the prognosis is not significantly affected by whether the disease is diagnosed prenatally or postnatally.[1,2]

The severity of the disease, pulmonary complications, long-term management, complications of surgical interventions, and outcomes need to be explained to the parents. The possibility of developing intrauterine heart failure and hydrops in absent pulmonary valve should also be explained to the parents. In our case, the above was explained to parents, and then, the pregnancy was terminated.

Further studies on this anomaly may be required concerning natural courses, the survival of the fetus, and medical and surgical management outcomes that influence the decision of medical pregnancy termination.[5]

CONCLUSION

The absent pulmonary valve is a rare congenital heart disease and often associated to TOF, but has an overall poor prognosis. When a pericardiac cystic lesion is seen in the fetus, this abnormality is an important differential diagnosis. Its unique features are the aneurysmal dilatation of the main pulmonary artery and branches and significant pulmonary valve regurgitation. The outcomes for prenatally diagnosis vary significantly depending on the underlying lesions.

The prenatal diagnosis of an absent pulmonary valve may be possible with a high degree of accuracy with fetal echocardiographic findings. Therefore, early fetal screening ultrasonography (and fetal echocardiography if there is any suspicion of disease) should be considered.

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