

# Association of Pulmonary Atresia with Intact Ventricular Septum and Aortic Valve Stenosis. Prenatal Diagnosis

Ivan Romero Rivera, Valdir Ambrósio Moises, Celia Camelo Silva, José Lázaro Andrade, Antonio Carlos Carvalho

São Paulo, SP - Brazil

*A rare association of pulmonary atresia with an intact septum was diagnosed through echocardiography in a fetus 32 weeks of gestational age. The diagnosis was later confirmed by echocardiography of the newborn infant and further on autopsy. The aortic valve was bicuspid with a pressure gradient of 81mmHg, and the right ventricle was hypoplastic, as were the pulmonary trunk and arteries, and the blood flow was totally dependent on the ductus arteriosus.*

Association of pulmonary atresia with intact ventricular septum and aortic valve stenosis is extremely rare with only a few cases reported in the literature<sup>1</sup>, and none in our country. We report one case of this rare association, diagnosed by fetal echocardiography and confirmed by transthoracic echocardiography of the newborn and further on autopsy.

## Case report

A 25-year-old woman in the 32<sup>nd</sup> week of gestation was referred for fetal echocardiographic study because of a suspicion of fetal cardiac malformation on an echocardiography performed two weeks earlier. The woman had no familial antecedents of heart disease and did not use teratogenic agents. This was her second pregnancy. Her first had been uneventful resulting in a normal delivery.

The fetal echocardiogram showed situs solitus, the heart and its apex located to the left of the thorax, and atrioventricular and ventriculoarterial concordance. The atria were moderately dilated and the color-flow mapping showed right-left laminar blood flow through the wide atrial septal defect of the ostium secundum type. The tricuspid valve was hypoplastic and competent allowing, however, blood flow passage during diastole. The normal-sized mitral valve was mildly thickened and had a moderate reflux. The right ventricle was significantly

hypoplastic and hypertrophied, and the ventricular inlet and outflow tract could be identified (fig. 1). The visualized body was totally obliterated with images suggesting intramyocardial sinusoids (fig. 2). The normal-sized left ventricle had significant symmetrical hypertrophy. The pulmonary valve was mildly thickened and showed no antegrade flow on the Doppler study and color-flow mapping. The pulmonary trunk had a diameter of 3mm and normal bifurcation (fig. 2). The bicuspid aortic valve was moderately thickened, showed a maximum systolic pressure gradient of 81mmHg, with no reflux, and a poststenotic dilation when the ascending aorta was visualized. The aortic arch was normal and the area of the ductus arteriosus was not visualized (fig. 3).

In the 37<sup>th</sup> week of gestation a normal delivery took place. The female newborn weighed 2,200g and showed an Apgar score of 6/8. The newborn was referred to the intensive care unit where she arrived pale and cyanotic. Pulmonary auscultation showed symmetric respiratory sounds and disseminated rales. On cardiac auscultation, the second cardiac sound was single and of diminished intensity, and an ejection cardiac murmur (+++) was heard on the high left sternal margin. The arterial saturation of oxygen was 65% at FIO<sub>2</sub> 0.3.

Chest X-ray showed a normal cardiac silhouette and bilateral pulmonary flow reduction. The electrocardiogram showed sinus rhythm, electric axis of ventricular activation at +60°, right atrial hypertrophy, absence of the right ventricular electrical potentials, and signs suggesting left ventricular ischemia. The echocardiogram confirmed the findings of the fetal echocardiogram. The aortic gradient, however, was 61mmHg and the ductus arteriosus was patent with no signs of obliteration.

The newborn received prostaglandin E1 and dopamine, and the possibility of surgical treatment to correct the aortic valve was discussed. Systemic-pulmonary derivation through Blalock-Taussig surgery or dilation of the aortic valve by balloon after hemodynamic stabilization were also considered. After several episodes of significant bradycardia and progressive cyanosis, however, the newborn died before any intervention. Autopsy confirmed the echocardiographic findings.

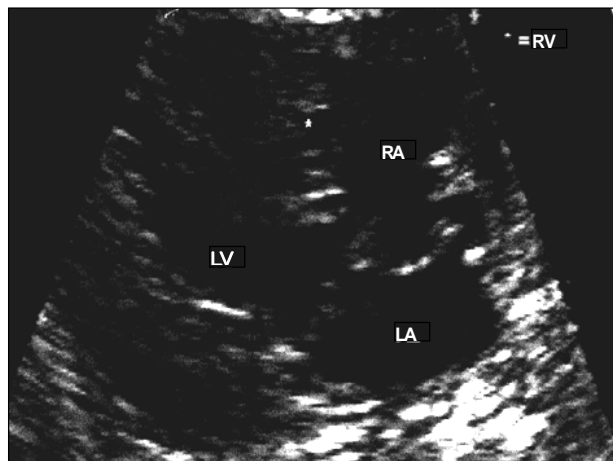


Fig. 1 – Four-chamber fetal echocardiographic image showing left ventricular hypertrophy and extreme right ventricular hypoplasia. RA- right atrium; RV- right ventricle; LA- left atrium; LV- left ventricle.

## Discussion

Among all cardiac malformations associated with pulmonary atresia with an intact ventricular septum, aortic

valve stenosis is probably the rarest with only a few reports of isolated cases in the literature<sup>1</sup>. Mortality is high in the reported cases even with surgical treatment, because significant hemodynamic alterations occur in the left ventricle. These alterations are partially due to the pressure overload resulting from the aortic valve stenosis, in some cases with clear subendocardial ischemia<sup>1,5</sup> on the electrocardiogram, and also partially due to the possibility of impairment of the coronary artery circulation that exists in pulmonary atresia with an intact septum<sup>1,6</sup>. In addition, the pulmonary circulation is totally dependent on left ventricular ejection in these cases, which also determines a significant volumetric overload. Prenatal diagnosis by fetal echocardiography makes this case even more interesting because we do not know of any similar reports, even in the international literature where isolated cases of prenatal diagnoses of tetralogy of Fallot with aortic valve stenosis have been reported<sup>2-4</sup>.

This complex case illustrates the diagnostic accuracy that can be obtained through fetal echocardiography, confirming once more the need for performing this important diagnostic procedure in all cases where suspicion of fetal cardiac malformation exists.

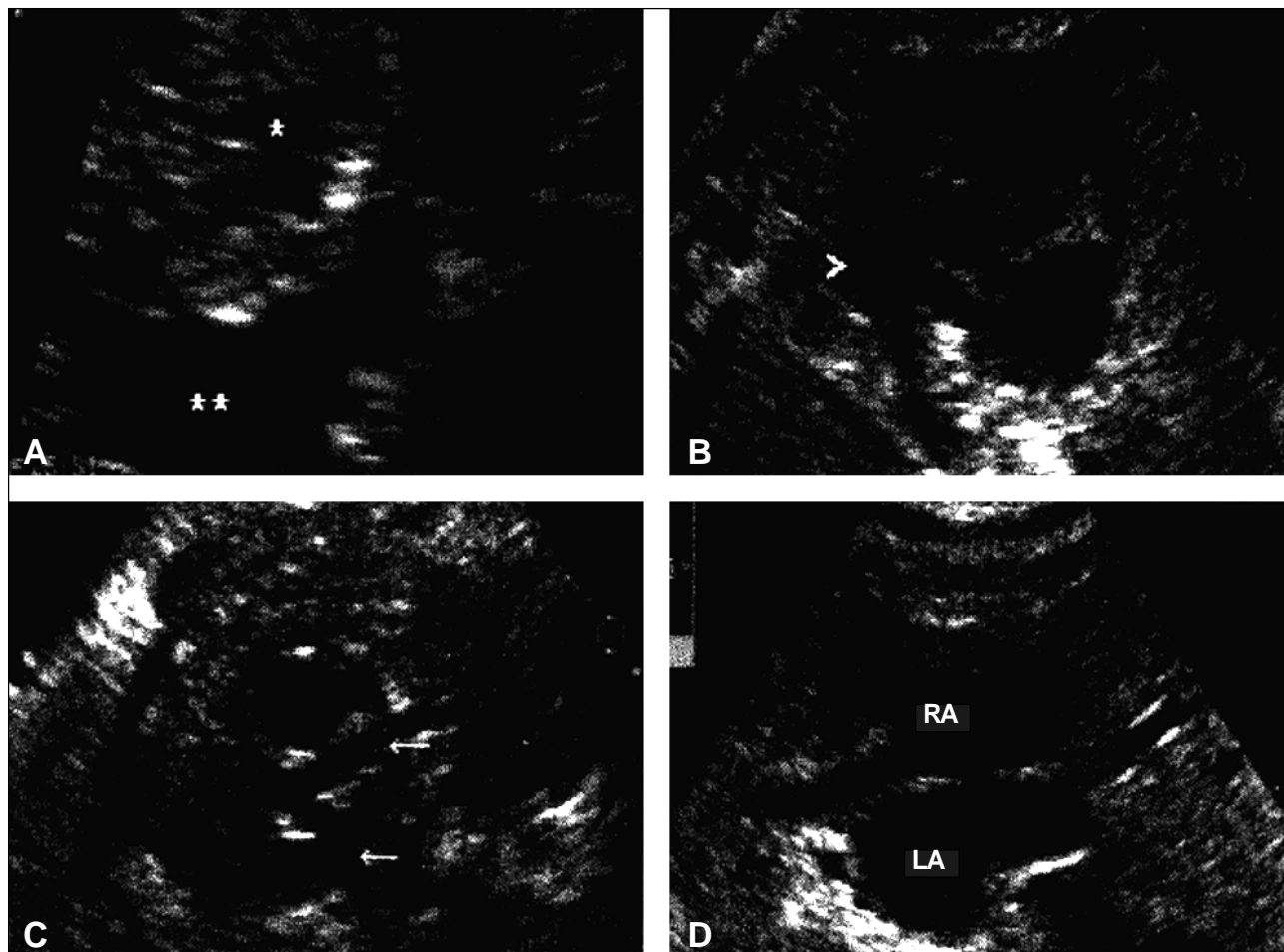


Fig. 2 – Fetal echocardiographic data showing: A) right ventricular hypoplasia (\*) with an image suggesting intramyocardial sinusoid. Note the left ventricular hypertrophy (\*\*); B) pulmonary trunk (arrow head); C) normal origin and trajectory of the pulmonary arteries (arrows); D) interatrial septum and also confluence of the venae cava with the right atrium. RA- right atrium; LA- left atrium.

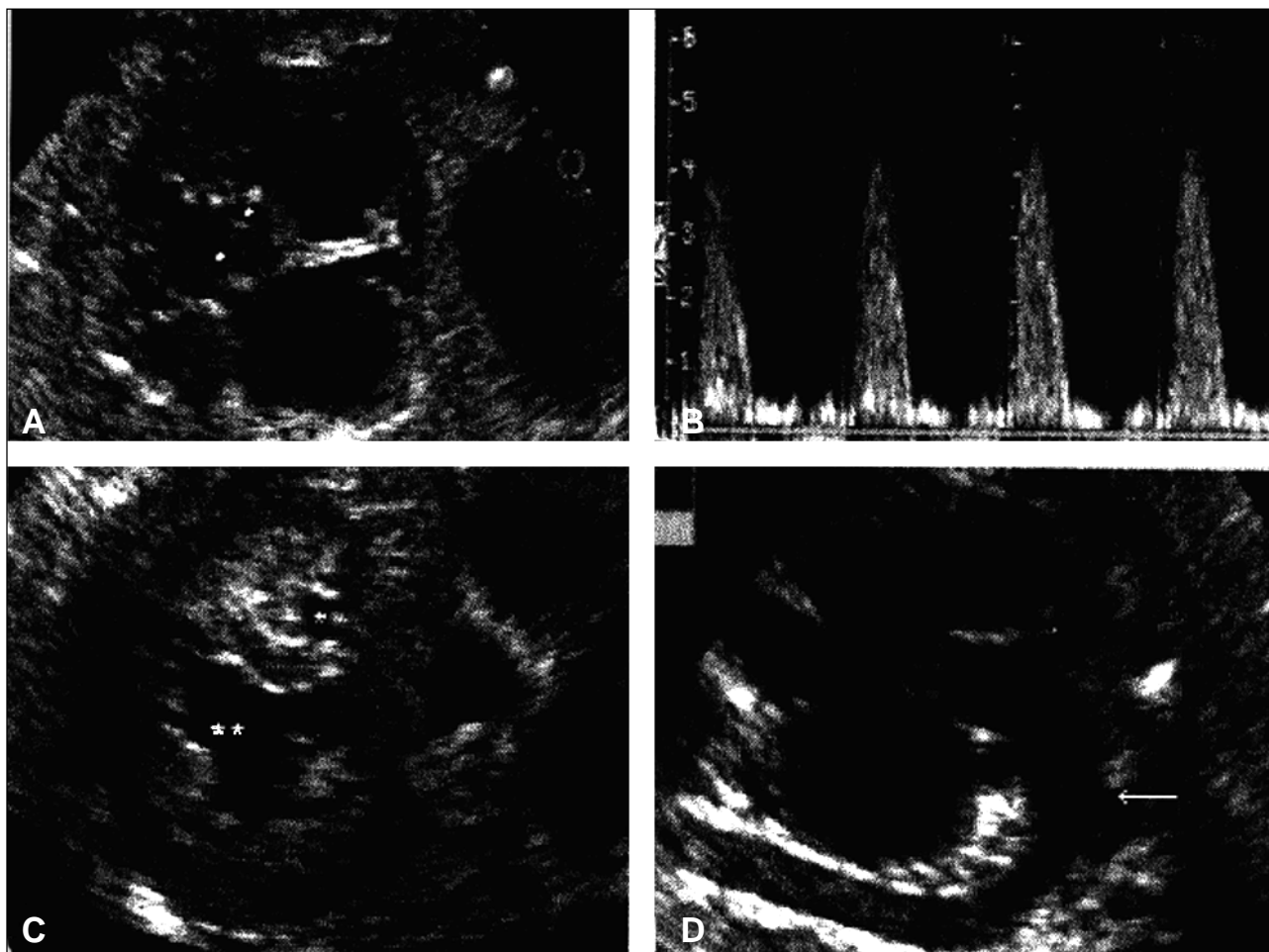


Fig. 3 – Fetal echocardiographic data showing: A) bicuspid aortic valve (\*=valve cusps); B) systolic gradient through the aortic valve; C) right ventricular hypoplasia (\*) and left ventricular hypertrophy (\*\*); D) normal aortic arch (arrow), with poststenotic dilation of the ascending aorta.

## References

1. Patel RG, Freedom RM, Bloom KR, Rowe RD. Truncal or aortic valve stenosis in functionally single arterial trunk: a clinical, hemodynamic and pathologic study of six cases. *Am J Cardiol* 1978; 42: 800-9.
2. Rowland DG, Caserta T, Foy P, Wheller JJ, Allen HD. Congenital absence of the pulmonary valve with tetralogy of Fallot with associated aortic stenosis and patent ductus arteriosus: a prenatal diagnosis. *Am Heart J* 1996; 132: 1075-7.
3. Salazar J. Prenatal diagnosis of tetralogy of Fallot with absent pulmonary valve and aortic stenosis. *Eur Obstet Gynecol Reprod Biol* 1998; 76: 11-3.
4. Ino T, Yabuta K, Okada R. Rare association of tetralogy of Fallot and aortic valve stenosis: autopsy findings after failed balloon aortic valvuloplasty. *Jpn Heart J* 1992; 33: 125-30.
5. Vincent WR, Buckberg GD, Hoffman JIE. Left ventricular subendocardial ischemia in severe valvar and supra-valvar aortic stenosis: a common mechanism. *Circulation* 1974; 49: 326-33.
6. Rivera IR, Moises VA, Silva CC, Abujamra P, Andrade JL, Carvalho AC. Circulação coronária dependente do ventrículo direito na atresia pulmonar com septo íntegro. Ausência da origem das artérias coronárias da aorta. *Arq Bras Card* 1998; 71: 143-6.