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TWO-DIMENSIONAL AND DOPPLER ECHOCARDIOGRAPHY IN THE DIAGNOSIS OF ABSENT PULMONARY VALVE SYNDROME (SURGERY WITHOUT CATHETERIZATION)*

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SUMMARY: Özkutlu S, Özme Ş, Atalay S, Saraçlar M, Bilgiç A, Yurdakul Y. (Department of Pediatric Cardiology, Hacettepe University Institute of Child Health, Ankara, Turkey). Two-dimensional and Doppler echocardiography in the diagnosis of absent pulmonary valve syndrome (surgery without catheterization). Turk J Pediatr 34: 245-250, 1992.

The majority of severely affected infants with absent pulmonary valve syndrome develop varying degrees of respiratory distress. Cardiac catheterization and angiocardiography are still performed in the diagnosis of these infants prior to surgery. However, considering the high risks of these invasive investigations for severely symptomatic infants, diagnosis using only noninvasive methods becomes important. In this regard, we present three cases of absent pulmonary valve syndrome, diagnosed pre-operatively by both two-dimensional and Doppler echocardiography. The diagnosis was confirmed by cardiac catheterization and angiocardiography in two cases and by surgery in a symptomatic infant. Therefore, we are of the opinion that the two-dimensional and Doppler echocardiograpic methods are most reliable in diagnosing absent pulmonary valve syndrome, and that severely symptomatic infants can be referred for surgery without catheterization. *Key words: two-dimensional echocardiography, Doppler echocardiography, absent pulmonary valve syndrome.*

Congenital absence of the pulmonary valve is a rare cardiac malformation usually associated with tetralogy of Fallot (TOF)[1-3]. Patients generally present in early infancy with cyanosis or respiratory distress or both[4]. Clinical findings may be complicated by respiratory insufficiency due to tracheobronchial compression by the dilated pulmonary arteries[4]. This syndrome has been diagnosed by cardiac catheterization and angiocardiography. In a few recent reports, it was emphasized that since echocardiographic features of absent pulmonary valve syndrome appear to be unique, diagnosis can be established by the use of noninvasive methods[5-7].

When considering the high risks involved for severely symptomatic infants undergoing cardiac catheterization, it is important that only noninvasive methods be used in arriving at a diagnosis before surgery is undertaken. To illustrate this

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point, we present three cases of congenital absence of the pulmonary valve diagnosed by both two-dimensional and Doppler echocardiography. The diagnosis was subsequently confirmed by cardiac catheterization, angiocardiography and cardiac surgery in two patients and by surgery in the severely symptomatic infant.

Material and Methods

Among the patients referred to the Department of Pediatric Cardiology of the Hacettepe University Institute of Child Health, three patients were diagnosed as having absent pulmonary valve syndrome by the use of both two-dimensional and Doppler echocardiography. In addition, cardiac catheterization and angiocardiography were performed in two patients. The ages of the patients ranged between two months and seven years. All patients underwent cardiac surgery.

The echocardiographic examination included M-mode, two-dimensional, contrast and Doppler studies using a Toshiba-Sonolayer-S SSH 60 A Scanner with 2.5, 3.75 and 5 MHz transducers.

Case Report

Clinical manifestations and laboratory findings of these patients are presented in Table I, and some echo findings in Table II. Other echo findings were as follows:

In all patients right ventricular enlargement and subaortic ventricular septal defect, also demonstrated by contrast studies, were noted. In the short-axis view at the level of the great arteries an echo-dense structure at the level of the pulmonary annulus, suggestive of valve tissue and dilated pulmonary arteries, was observed

Table I: Clinical and Laboratory Data of Patients

Case No.	Age Sex	Symptoms	Murmur	ECG	X-ray	Pressure (mm Hg)	Angiocardiography
1	4 yr. M	No	To-and-fro	Right axis RV Hypertrophy	Cardiomegaly	RVP : 110/5 PA : 28/9	Dilated main PA and branches, VSD PR
2	7 yr. M	No	To-and-fro	RV Hypertrophy	Cardiomegaly Pulmonary Pressure	RVP : 110/5 PA : 35/4	Dilated main PA and branches Narrow PA annulus VSD PR
3	2 Mo. F	Dyspnea Cough Cyanosis	To-and-fro	Right axis RV Hypertrophy	Cardiomegaly Prominent RPA		

RV : right ventricle
RPA : right pulmonary artery
PA : pulmonary artery
PR : pulmonary regurgitation

Table II. Echo Findings

Case No.	Diameter of PA Annulus (mm)	Diameter of Main PA (mm)	Diameter of LPA (mm)	Diameter of RPA (mm)	Diameter of Ao Annulus (mm)	Systolic Gradient By Doppler Echo (mm Hg)
1	12	30	27	17	20	75
2	15	46	17	22	22	67
3	8	14	12	18	11	68

PA : pulmonary artery
LPA : left pulmonary artery
Ao : aorta

in each patient. In the subcostal position, the right and left pulmonary artery branches were visualized more clearly (Fig. 1).

In the parasternal short-axis view, retrograde systolic and anterograde diastolic flow patterns in the main pulmonary artery were recorded by continuous-wave Doppler echocardiography (Fig. 2). Diameters of the pulmonary arteries and the systolic gradient across the pulmonary annulus measured by Doppler echocardiography are noted in Table I.

Surgical confirmation of the diagnosis was available in all three patients.

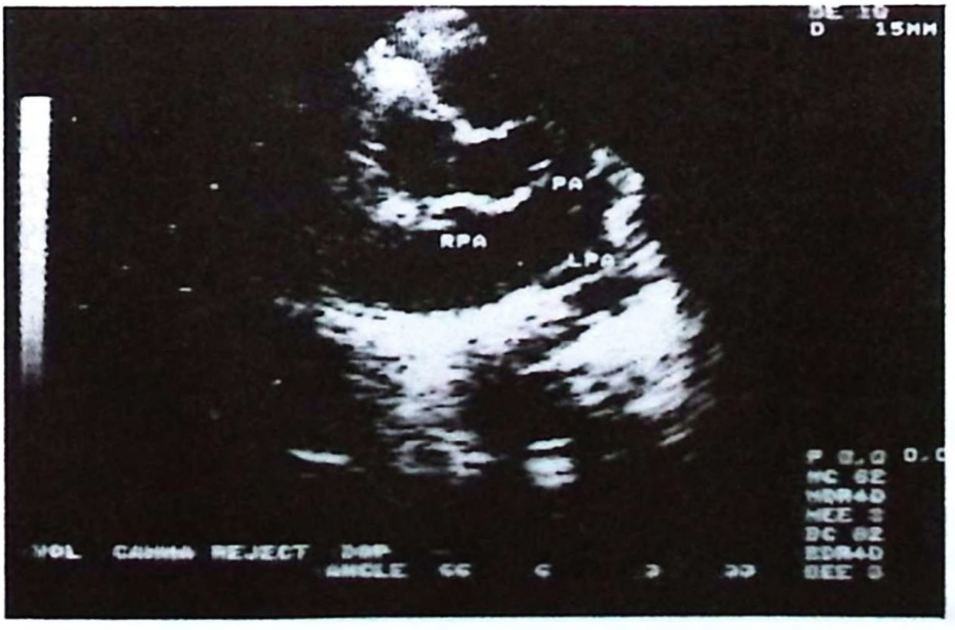

Fig. 1: Case 3. Two-dimensional echocardiogram in the subcostal position. The main and right pulmonary arteries are markedly dilated. (PA: pulmonary artery, RA: right pulmonary artery, LA: left pulmonary artery).

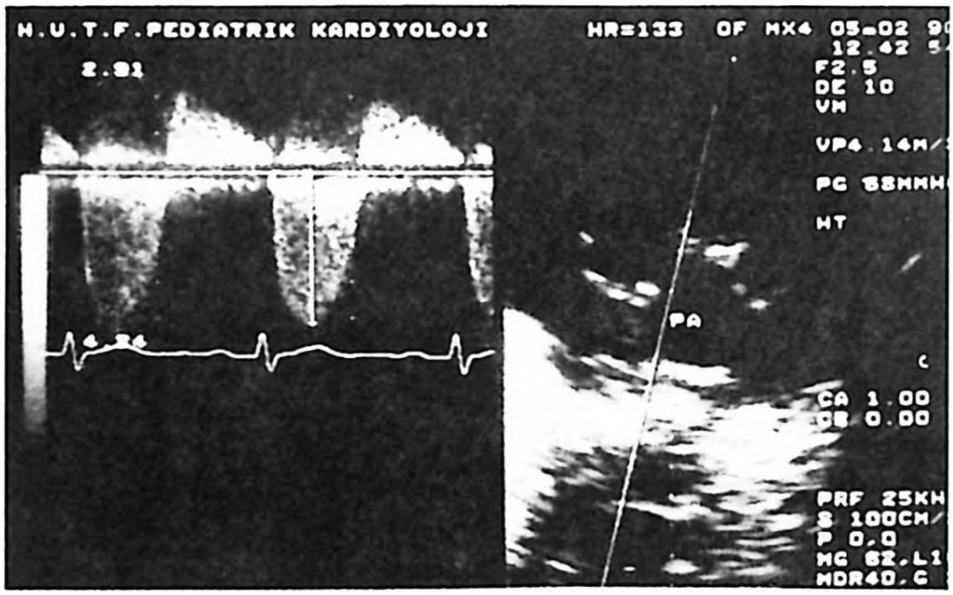

Fig. 2: Same patient. Two-dimensional and continuous-wave Doppler echocardiogram in the parasternal short-axis view showing antegrade systolic flow (below the baseline) and retrograde diastolic flow (above the baseline) in the main pulmonary artery.

Discussion

The absent pulmonary valve syndrome was first described by Chevers in 1847[8]. This anomaly can be associated with other congenital anomalies such as TOF, ventricular septal defect (VSD), patent ductus arteriosus (PDA), atrial septal defect (ASD), double outlet right ventricle (DORV), endocardial cushion defects (ECD), tricuspid atresia and transposition of the great arteries (TGA)[4]. It is rarely present as an isolated lesion[4]. The cases presented were associated with VSD. Only 15 cases without VSD have been reported in the literature[4].

In most of our patients, symptoms of respiratory distress were present in infancy and early childhood due to the compression of the trachea by a pulmonary artery aneurysm. While two cases were asymptomatic and were referred to our hospital for evaluation of a to-and-fro murmur detected on physical examination, the third had cyanosis and dyspnea.

There are reports of a few patients who have been diagnosed only by two-dimensional echocardiography or by both two-dimensional and Doppler echocardiography[5-7]. Ventricular septal defect, aortic overriding and right ventricular dilatation can be demonstrated by two-dimensional echocardiography. In the parasternal short-axis view of the great arteries, main pulmonary artery dilatation and dysplastic or rudimentary pulmonary valve elements can be recorded[4-7]. In the cases presented, normal septal-aortic continuity, VSD and right ventricular dilatation were seen. In all three cases, it was demonstrated in the short-axis view of the great arteries, that the main pulmonary artery was dilated, the annulus was

stenotic and the pulmonary valves were rudimentary. In the subcostal position, branches of the right and left pulmonary arteries were visualized far more clearly.

With Doppler echocardiography in the parasternal short-axis view, a retrograde systolic flow pattern was recorded in the main pulmonary artery and antegrade diastolic flow patterns in the main pulmonary artery and in the right ventricular outflow tract.

The distinction between the resemblance of the Doppler flow pattern in the main pulmonary artery in absent pulmonary valve syndrome and the flow pattern in PDA can be made in the absence of a diastolic flow at the right ventricular outflow tract unless pulmonary insufficiency in association with PDA is present[9].

Since 1982, neonates and infants with certain types of congenital heart disease such as critical aortic valve stenosis, pulmonary atresia or stenosis, aortic arch obstruction, aortic atresia, total anomalous pulmonary venous connection, and truncus arteriosus are referred for surgery only after echocardiographic evaluations are made. Of course, this decision depends upon the confidence and skill of the referring cardiologist[10,11]. When considering the high risks involved in catheterization of symptomatic newborns and small infants, the importance of referring these patients for surgery without undergoing catheterization increases.

Therefore, it is our opinion that the two-dimensional and Doppler echocardiographic methods are most reliable for diagnosing absent pulmonary valve syndrome and that severely symptomatic infants may undergo surgery without catheterization, thus avoiding the high risks of this invasive procedure.

REFERENCES

1. Fischer DR, Neches WH, Beerman LB, et al. Tetralogy of Fallot with absent pulmonic valve: analysis of 17 patients. *Am J Cardiol* **53**: 1433, 1984.
2. Ilbawi MN, Fedorchik J, Muster AJ, et al. Surgical approach to severely symptomatic newborn infants with tetralogy of Fallot and absent pulmonary valve. *J Thorac Cardiovasc Surg* **91**: 584, 1986.
3. Bilgiç A, Saraçlar M, Özme Ş, et al. The absent pulmonary valve syndrome. *Turk J Pediatr* **23**: 121, 1981.
4. Emmanouilides GC, Doroshow RW. Congenital absence of the pulmonary valve. In Adams FH, Emmanouilides GC, Riemenschneider TA (eds). *Moss' Heart Disease in Infants, Children, and Adolescents* (4th ed). Baltimore: Williams and Wilkins Co, 1989, pp. 301-307.
5. Cheatham JP, Latson LA, Gutgesell HP. Echocardiographic pulsed Doppler features of absent pulmonary valve syndrome in the neonate. *Am J Cardiol* **49**: 1773, 1982.
6. Di Segni E, Einzig S, Bass JL, Edwards JE. Congenital absence of pulmonary valve associated with tetralogy of Fallot: diagnosis by 2-dimensional echocardiography. *Am J Cardiol* **51**: 1798, 1983.
7. Tenorio de Albuquerque AM, Ortiz J, Villela de Moraes A, et al. Cross-sectional echocardiographic features of 'absent' pulmonary valve. Report of two cases. *Int J Cardiol* **5**: 155, 1984.

8. Chevers N. Recherches sur les maladies de l'artere pulmonaire. *Arch Gen Med* 15: 488, 1847.
9. Stevenson JG, Kawabori I, Guntheroth WG. Pulsed Doppler echocardiographic diagnosis of patent ductus arteriosus: sensitivity, specificity, limitations, and technical features. *Cathet Cardiovasc Diagn* 6: 255, 1980.
10. Huhta JC. Heart surgery without catheterization. In Adams FH, Emmanouilides GC, Riemenschneider TA (eds). *Moss' Heart Disease in Infants, Children, and Adolescents* (4th ed). Baltimore: Williams and Wilkins Co, 1989, pp. 996-1003.
11. Saraçlar M, Özme Ş, Özkutlu S, et al. Preoperative evaluation of congenital heart disease by echocardiography. *19th International Congress of Pediatrics Abstracts*. Paris, p. 416, 1989.