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ABSENT PULMONARY VALVE SYNDROME WITH AGENESIS OF THE LEFT PULMONARY ARTERY*

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SUMMARY: Paç A, Özme Ş, Çeliker A, Özkutlu S. (Cardiology Unit, Department of Pediatrics, Hacettepe University Faculty of Medicine, Ankara Turkey). Absent pulmonary valve syndrome with agenesis of the left pulmonary artery. Turk J Pediatr 1994; 36: 249-253.

Absent pulmonary valve syndrome is represented by rudimentary nodules without identifiable leaflets, and it is commonly associated with a ventricular septal defect and tetralogy of Fallot. Unilateral absence of a pulmonary artery is also a rare congenital anomaly. The following is a case report of a one-day-old newborn with cyanosis. The physical examination revealed a to-and-fro murmur and no expansion of the left hemithorax. Echocardiography revealed tetralogy of Fallot, absent pulmonary valve, and enlarged main and right pulmonary arteries. The catheterization and angiography confirmed the diagnosis and absence of the left pulmonary artery. *Key words: absent pulmonary valve syndrome, tetralogy of Fallot, pulmonary artery agenesis.*

Absent pulmonary valve syndrome is represented by a complete absence of pulmonary valve leaflets or an uneven rim of rudimentary tissue found in the area of the pulmonary annulus. There is also stenosis of the pulmonary annulus. The most often recognized characteristic of the syndrome is aneurysmal dilatation of the pulmonary trunk and one or both of its branches[1].

This syndrome can occur as an isolated lesion and is also commonly associated with ventricular septal defect and tetralogy of Fallot[1-4]. Other malformations such as atrial septal defect, double-outlet right ventricle, patent ductus arteriosus, endocardial cushion defect, transposition of the great arteries and other complex cardiac malformations have occasionally been described in association with absent pulmonary valve syndrome[1,4,5].

On the other hand, congenital unilateral absence of a pulmonary artery is also a rare anomaly which is frequently associated with other cardiovascular abnormalities such as tetralogy of Fallot[6].

These two congenital anomalies detected together are presented because of their very rare occurrence.

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Case Report

A one-day-old female newborn was admitted to the hospital in December 1991 with the complaint of cyanosis. Birth weight was 3300 g. A marked left parasternal impulse and to-and-fro systolic and diastolic murmurs were noted along the left upper sternal border. The second heart sound was single and increased in intensity in the second left intercostal space. No expansion was seen in the left hemithorax. Auscultation revealed no sound in the left lung. The electrocardiogram showed sinus rhythm, right axis deviation and right ventricular hypertrophy. The chest radiograph revealed a homogeneously dense left hemithorax (Fig. 1). The echocardiography showed an overriding aorta with a subaortic ventricular septal defect and a markedly dilated main pulmonary artery and right pulmonary artery. Pulmonary annulus was notably narrow, and a thickened rim of tissue was found in the region of the pulmonary valve annulus (Fig. 2).

Doppler echocardiography demonstrated a 45 mm Hg systolic gradient and diastolic retrograde flow at the pulmonary valve level. The left pulmonary artery could not be detected by two-dimensional echocardiography. The patient

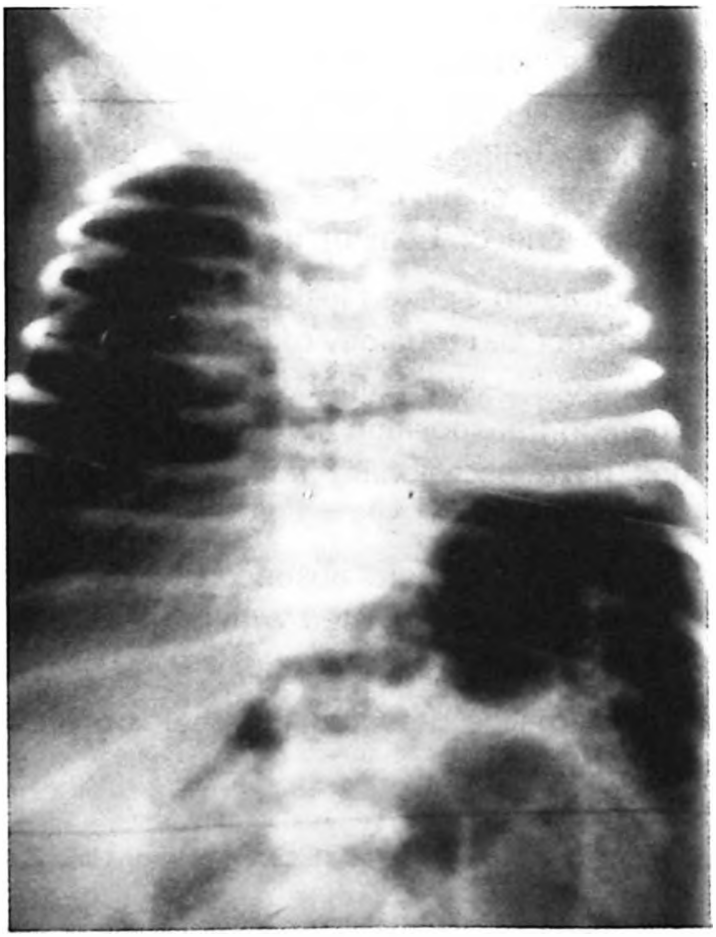

Fig. 1: The chest roentgenogram of the patient showing dense left hemithorax.

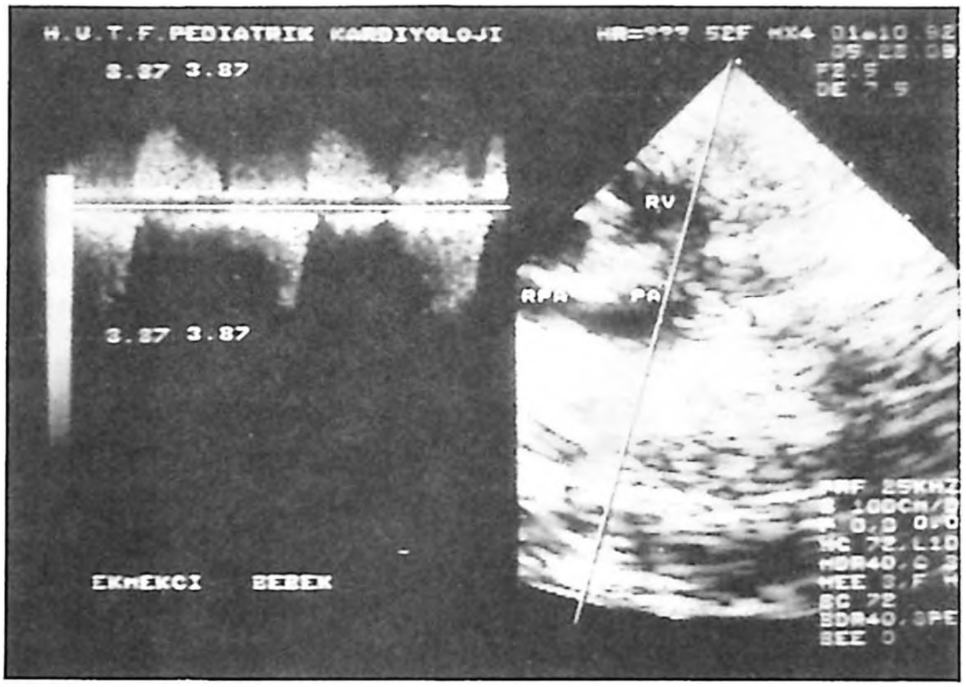

Fig. 2: The echocardiographic appearance of the patient. It shows enlarged main and right pulmonary arteries and absence of the left pulmonary artery. Doppler echocardiography demonstrates systolic flow with an important gradient across the right ventricular outflow tract and diastolic flow.

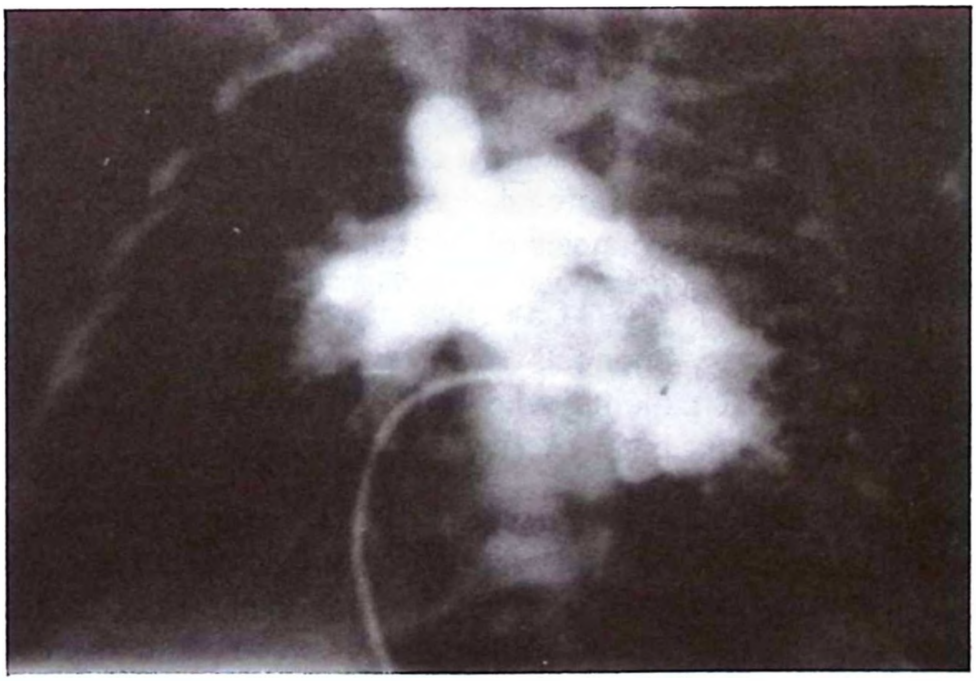

Fig. 3: Angiography of the right ventricle, revealing overriding aorta, right-sided aortic arch, dilated main and right pulmonary arteries, and absence of the left pulmonary artery.

underwent cardiac catheterization, which confirmed the diagnosis of absent pulmonary valve syndrome and detected the absence of the left pulmonary artery.

Hemodynamic findings of right-side cardiac catheterization were as follows: right ventricular systolic pressure 45 mm Hg, and main pulmonary artery systolic/diastolic pressure 16/2 mm Hg. Angiography revealed a ventricular septal defect, overriding aorta, enlarged main and right pulmonary arteries, right-sided aortic arch, and absence of the left pulmonary artery. Regurgitant flow was detected at the pulmonary valve level (Fig. 3).

Respiratory distress was not seen during the patient's ten-day hospital stay, and she was discharged. She is still being followed by the Cardiology Department.

Discussion

Absent pulmonary valve syndrome is seen in two well-defined groups. The first group comprises infants with severe respiratory symptoms in the neonatal period. Unfortunately, most infants who are symptomatic die as a result of heart failure or bronchial compression[7]. Morbidity and mortality in this group are high[7-9]. The second group comprises older children whose symptoms are less severe. Although our patient was a one-day-old newborn, she had no respiratory distress. Respiratory failure is most often due to tracheobronchial compression by the massively enlarged pulmonary arteries, and treatment has to date focused on reduction of the pulmonary arterial size[7].

Absence of the pulmonary valve is usually associated with a ventricular septal defect, significant pulmonic stenosis and a absent ductus arteriosus[7]. It has been postulated that closure of the arterial duct in early intrauterine life plays an important etiologic role in the genesis of this syndrome[10,11]. In our case as well, the arterial duct was closed.

Unilateral pulmonary artery agenesis is an anomaly uncommonly seen in the general population. Most patients who have pulmonary artery agenesis have abnormal chest x-ray films but lack respiratory symptoms.

Absent pulmonary valve syndrome can be diagnosed accurately with two-dimensional echocardiography[1,12]. There are, however, certain limitations to this technique, and evaluation of peripheral and intraparenchymal pulmonary arteries is one of them. We diagnosed absent pulmonary valve syndrome in our case by echocardiography, but the left pulmonary artery could not be seen. Therefore, the differential diagnosis was made by cardiac catheterization. We performed cardiac catheterization and confirmed the absent pulmonary valve syndrome. The left pulmonary artery was absent, and no vascular imaging was seen in the left hemithorax. Abnormalities of hilar branches of the pulmonary artery in absent

pulmonary valve syndrome were demonstrated on postmortem pulmonary angiography by Rabinovitch et al[13].

Unilateral absence of a pulmonary artery is a rare congenital anomaly that affects the right pulmonary artery more commonly than the left[6]. Typically the absent artery is on the side opposite the aortic arch, as seen in our patient. Tetralogy of Fallot is the most common underlying cardiac anomaly in patients with absence of the left pulmonary artery.

This is the first reported case of left pulmonary artery and parenchymal agenesis in absent pulmonary valve syndrome according to our knowledge of the literature in the last ten years.

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