The Turkish Journal of Pediatrics
Volume 23, No. 2, April - June 1981

THE ABSENT PULMONARY VALVE SYNDROME*

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Key words: absent pulmonary valve syndrome, congenital heart disease

The absence of a pulmonary valve is a rare congenital cardiac anomaly that may occur as an isolated lesion or, more commonly, in combination with other malformations of the heart. The combined form consists of severely hypoplastic pulmonary valve with annular stenosis, aneurysmal dilatation of the main pulmonary artery with one or both pulmonary branches also dilated, and a ventricular septal defect. In general, the term used in the literature for this combined anomaly has been tetralogy of Fallot with absent pulmonary valve[1-9]. However, Pinsky[10] recently reported this combination as a separate syndrome instead of as a variation of tetralogy of Fallot. The purpose of this report is to evaluate our cases from this standpoint and to describe the clinical, hemodynamic and angiographic findings of our nine cases.

Material and Methods

Of the nine cases studied 6 were males and 3 females. Their age range was from 2 days to 17 years.

All the patients were cyanotic at birth or shortly after birth, but only 2 patients remained cyanotic at rest after early infancy (Table I).

The physical findings were similar in all 9 cases. A prominent left parasternal impulse suggestive of right ventricular hypertrophy was present in all. There was a grade III - V/VI harsh basal systolic ejection murmur which radiated down the left parasternal border. The second heart sound was single. A prominent, low-frequency decrescendo early diastolic murmur was present in the second and third

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interspace at the left sternal border. The combination of a systolic ejection murmur and a rough early diastolic murmur resulted in a characteristic to-and-fro quality (Figure 1).

TABLE I

CLINICAL FINDINGS IN NINE CASES OF ABSENT PULMONARY VALVE SYNDROME

Case No.	Sex	Age at catheterization	Cyanosis At birth	Cyanosis Later	Respiratory distress	Pulmonary infection	Heart failure
1	M	2 days	+	With exertion	+	—	+
2	M	3 1/2 mos	+	None	+	+	—
3	M	12 mos	+	With exertion	+	+	+
4	M	4 mos	+	At rest	+	+	+
5	F	9 yrs	+	None	—	—	—
6	F	4 yrs	+	At rest	+	+	+
7	M	3 yrs	+	None	—	—	—
8	M	4 yrs	+	None	—	—	—
9	F	17 yrs	+	None	—	—	—

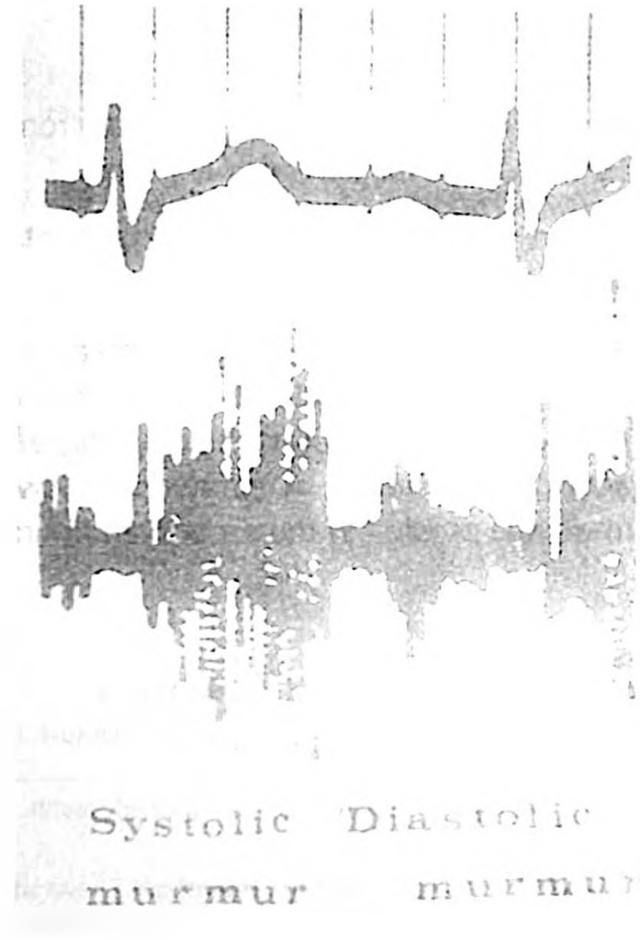

Figure 1

Case 8, Phonocardiogram showing the typical to-and-fro murmur.

The electrocardiographic features were consistent in all, showing right axis deviation and right ventricular hypertrophy (Figure 2).

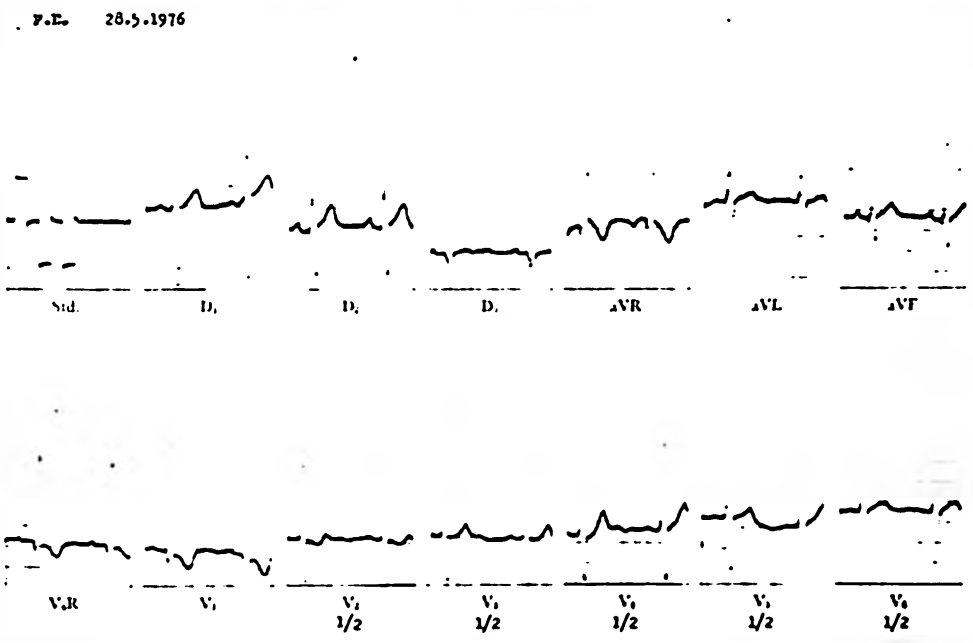

Figure 2.

Case 6, Electrocardiogram showing right axis deviation and right ventricular hypertrophy.

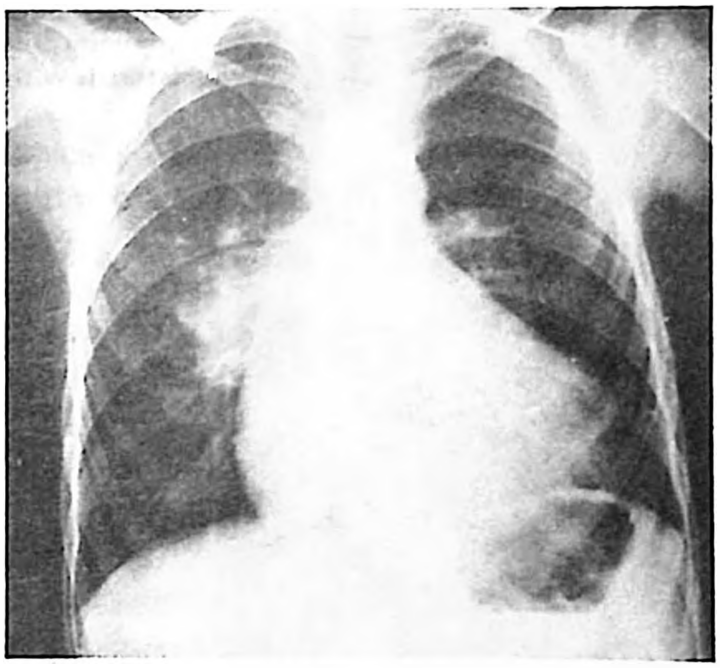

Figure 3.

Plain chest x-ray of case 5. The transverse diameter of the heart is increased. The right and left pulmonary arteries are aneurysmally dilated. The peripheral pulmonary vasculature is somewhat increased.

On their chest x-rays, the postero-anterior view demonstrated a moderately enlarged heart with massively dilated main, right and/or left pulmonary arteries (Figures 3-5). The pulmonary vasculature was increased in 4 cases, normal in 4 cases and decreased in one case. Three of the 9 cases had roentgenographic evidence of unilateral or bilateral emphysema. There was mediastinal shift in one case.

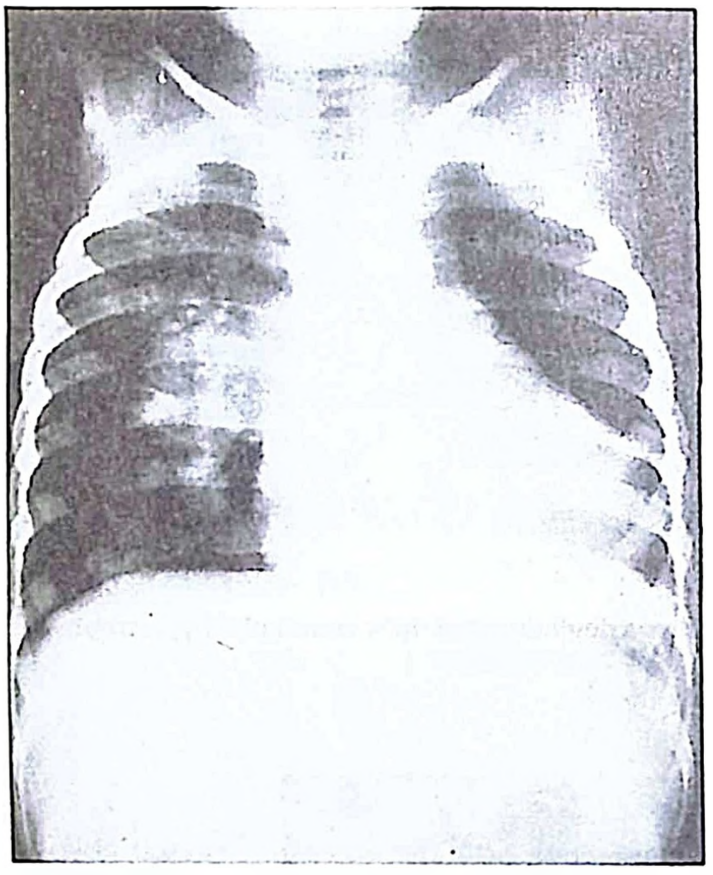

Figure 4

Case 6, Plain chest x-ray showing moderately enlarged heart and aneurysmally dilated right pulmonary artery. The peripheral pulmonary vasculature is within normal limits.

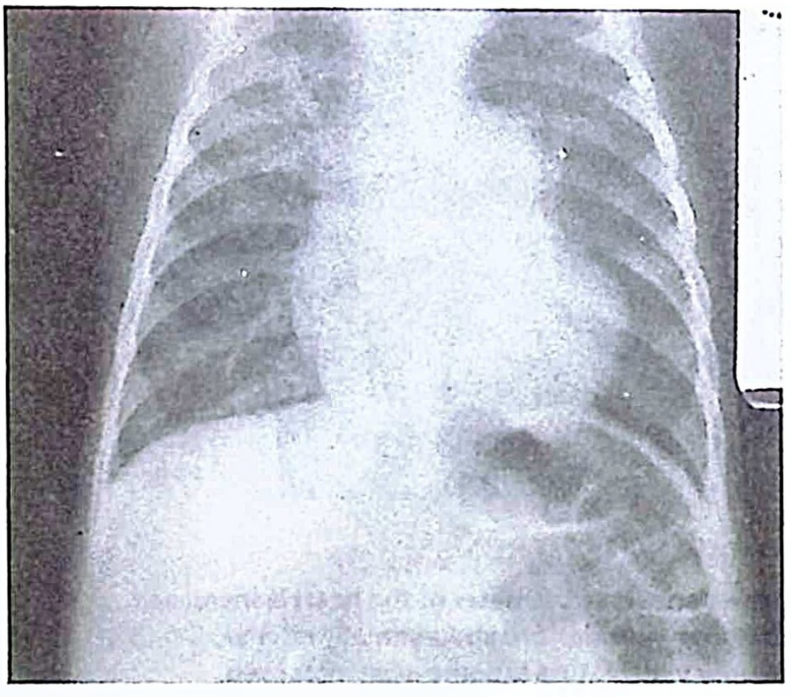

Figure 5

Plain chest x-ray, case 3. The main and left pulmonary arteries are aneurysmally dilated, and the peripheral pulmonary vasculature is increased.

At cardiac catheterization there was left-to-right shunt in 7 cases and right-to-left shunt in two cases. The peak systolic gradients across the pulmonary valve varied from 33 to 80 mmHg. Only two cases had infundibular stenosis (Table II).

TABLE II

CATHETERIZATION DATA ON NINE PATIENTS WITH ABSENT PULMONARY VALVE SYNDROME

Case No.	Pulmonary artery dilatation			Pressures (mmHg)			Infundibular stenosis	Dominant shunt
	max.	med.	min.	P A	R V	RV - PA		
1	MPA, LPA, RPA			—	75/0	—	—	L — R
2	RPA, MPA, LPA			27/10	60/0	33	—	R — L
3	MPA, RPA, LPA			20/6	75/0	55	+	L — R
4	RPA, MPA, LPA			40/10	80/0	40	—	R — L
5	MPA, RPA, LPA			22/5	95/0	73	—	L — R
6	RPA, MPA, LPA			30/10	90/0	60	—	L — R
7	RPA, MPA, LPA			20/5	100/0	80	—	L — R
8	MPA, RPA, LPA			22/10	90/8	68	—	L — R
9	RPA, MPA, LPA			30/15	100/-10	70	+	L — R

MPA = main pulmonary artery.
LPA = left pulmonary artery.
RPA = right pulmonary artery.

Cineangiocardiographically, all cases had pulmonary artery aneurysms that extended into the right and left main branches. Usually one side was affected more than the other, and this was commonly the right pulmonary artery (Figures 6, 7). The outflow tract stenosis was at the level of the pulmonary valve annulus where a discrete ridge of tissue was present. Valve leaflets could not be identified. In the two cases with infundibular stenosis, the entire infundibulum was directed from left to right and posteriorly (Figure 8).

Discussion

In 1847, Cheevers[11] reported a syndrome of congenital heart defects which included absent pulmonary valve, ventricular septal defect, annular pulmonary stenosis and dilatation of the main pulmonary artery and/or both pulmonary branches. In 1927 Kurtz, Sprague and White[12] described the physical findings and clinical course of a patient who at necropsy was found to have a ventricular septal defect, stenosis at the pulmonary valve ring and an absent pulmonary valve. According to Emmanoulides et al[4], approximately 150 such cases had been reported up to 1976.

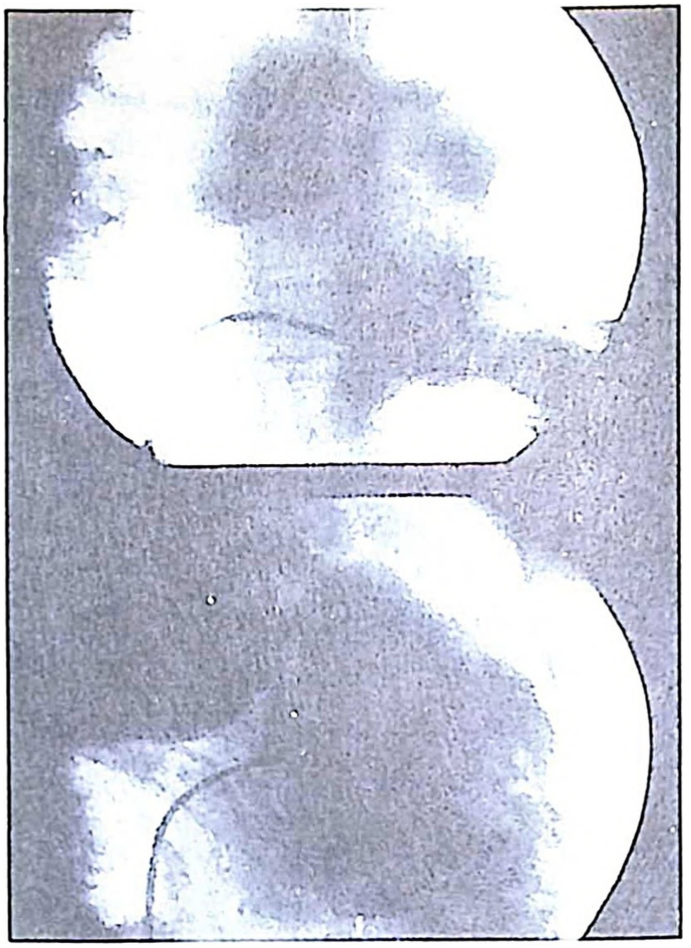

Figure 6

Cineangiocardiogram of case 5 (Top : antero-posterior view; bottom : right-anterior oblique view). Showing aneurysmally dilated right and left pulmonary arteries.

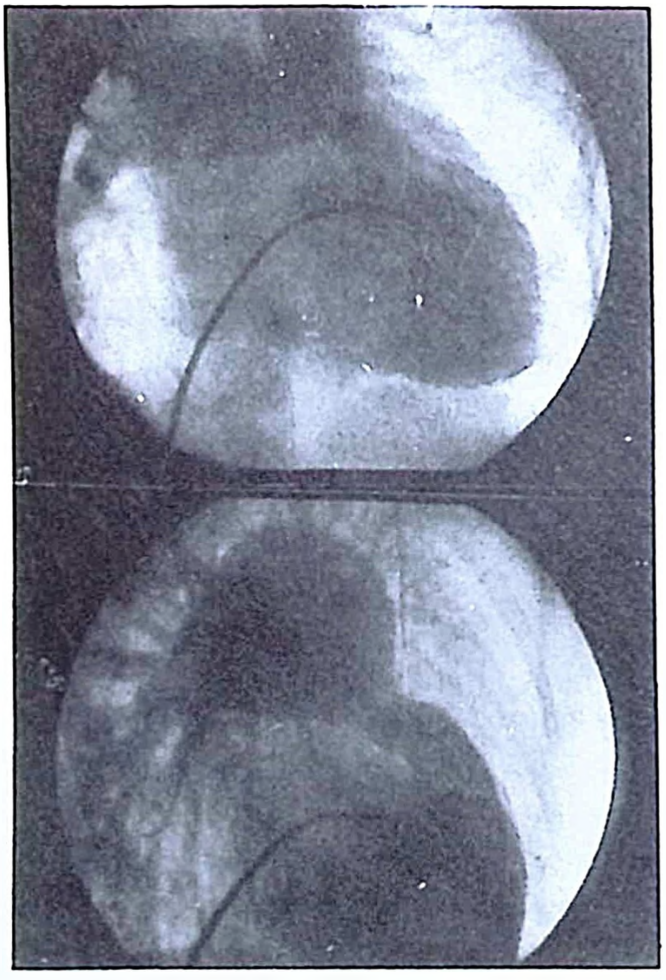

Figure 7

Cineangiocardiogram of case 7 (Top : antero-posterior view; bottom : right-anterior oblique view) Showing aneurysmally dilated right pulmonary artery.

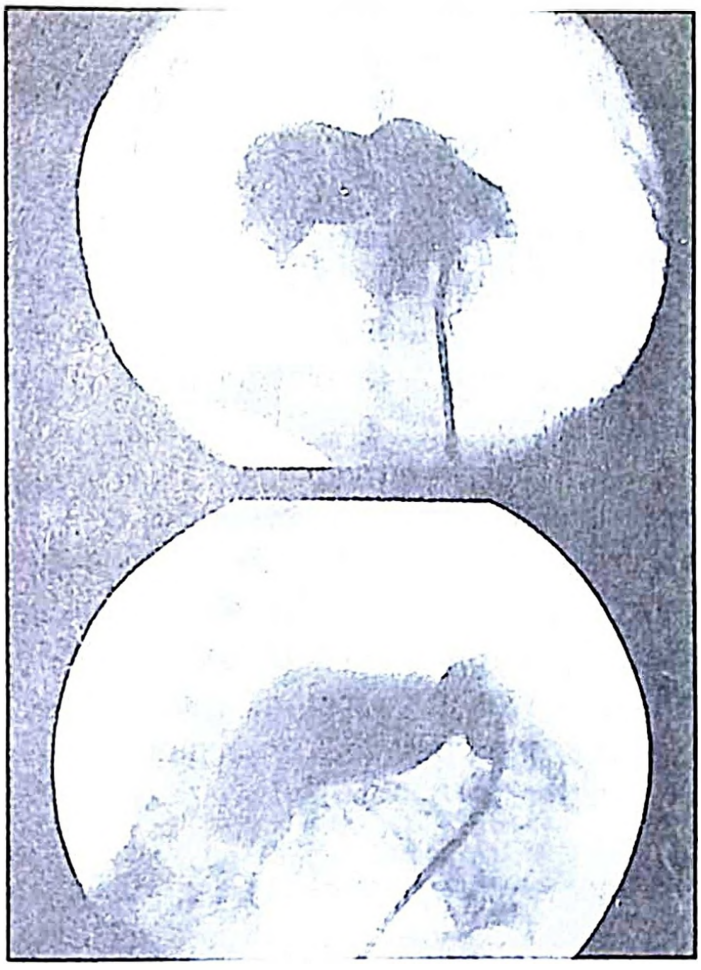

Figure 8

Cineangiocardiogram of case 4 (Top : antero-posterior view; bottom: right-anterior oblique view). Showing aneurysmally dilated right pulmonary artery, enlarged infundibulum and hypoplastic and stenotic pulmonary valve ring.

Infancy is the most critical period for patients with the absent pulmonary valve syndrome. Most of the symptoms appear to be respiratory in origin due to infection and airway obstruction secondary to the bronchial compression caused by dilated pulmonary arteries. Bronchial compression by distended pulmonary arteries may improve spontaneously towards the end of the first year. Several factors account for this improvement, and these are related primarily to maturational changes in the tracheobronchial tree and the lungs[5]. In our series 5 patients had respiratory distress in infancy. Recurrent respiratory infections, all beginning in infancy, were a prominent feature in 4 of the 9 cases. Congestive cardiac failure occurred in 4 patients during infancy (Table I). Three infants (aged 1 month to 7 months) died on initial or subsequent admissions, despite intensive therapy.

Little attention has been directed to the course of cyanosis in these patients. It is of considerable interest that although cyanosis was usually present in the neonatal period or infancy, almost invariably arterial oxygen saturation after this period was found to be above 89%, except in cases with severe pulmonary disease[5]. The disappearance of cyanosis and the occurrence of predominantly left-to-right shunts is probably the result of the decrease in pulmonary vascular resistance, which normally occurs in the neonatal period, and the improvement of pulmonary dysfunction with the maturation of the tracheobronchial tree. In our series only 2 of the 9 cases had cyanosis at rest, and the others were acyanotic.

A variety of palliative procedures have been performed in an attempt to relieve the pulmonary problems in infants. These are plication of aneurysmal pulmonary arteries[13], a sling around the dilated pulmonary artery attached to the retrosternal fascia[6], resection of the involved pulmonary artery with graft replacement[7], and resection of emphysematous lobes[8]. These palliative measures carry a high risk of mortality reaching 41%. Pinsky et al[10] recommend vigorous continual respiratory therapy for infants and small children and intracardiac repair in older children, since repair in infancy does not relieve respiratory symptoms.

Since infundibular stenosis is rare in the absent pulmonary artery syndrome, Lakier et al[5] recommend closing the ventricular septal defect as the primary procedure in corrective surgery. Two of our patients were operated on at age 8 and 9. Their ventricular septal defects were closed with a dacron patch. Dilatation of the pulmonary valve ring, with infundibular resection in one case, was performed. Despite resuscitative measures the latter patient died soon after the operation. Four patients are waiting for surgery to repair their intracardiac anomalies.

The term generally used for this combined anomaly has been "tetralogy of Fallot with absent pulmonary valve". However, in 1978 Pinsky reported it as a separate syndrome and suggested as a new term "the absent pulmonary valve syndrome". Although the new terminology has not gained wide acceptance, we prefer to evaluate our cases from that standpoint for the following reasons.

1. Only 2 of our 9 cases had cyanosis at rest. Stafford et al[9] reported one cyanotic case among 18 patients, and only one of Lakier's[5] 8 cases had cyanosis. The degree of cyanosis decreased with age, as was the case with our patients. This is in contradiction with the hemodynamics of the tetralogy of Fallot, where the cyanosis usually increases with age.
2. The problems during infancy were respiratory in origin, instead of being cardiac as in cases with tetralogy.
3. As in Lakier's cases, most of our patients showed dilated right ventricular infundibulum located superiorly and posteriorly. This is also different from the morphology of the infundibulum seen in the tetralogy of Fallot.
4. The pulmonary arteries and their branches were aneurysmally dilated instead of showing the pulmonary hypoplasia expected in the tetralogy of Fallot.

In conclusion, we believe that it would not be proper to term this anomaly "tetralogy of Fallot with absent pulmonary valve". We agree with Pinsky that the anomaly is more appropriately called "the absent pulmonary valve syndrome". However, further observations are needed.

Summary

The clinical, radiological, electrocardiographic and hemodynamic findings in 9 patients with the absent pulmonary valve syndrome are reported, and the relation of this syndrome to tetralogy of Fallot is discussed.

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