

Unilateral Absence of a Pulmonary Artery in Absent Pulmonary Valve Syndrome: A Case Report and Review of Literature

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A six-year-old boy presented to the Cardiology clinic with history of mild cyanosis and dyspnea on exertion from age 1. He had a to-and-fro murmur at the middle left sternal border. Chest examination was normal but chest x-ray showed a small left lung. Echocardiography established the diagnosis of tetralogy of Fallot (TOF) and absent pulmonary valve with severe pulmonary regurgitation and moderate stenosis at the pulmonary valve site. There was severe dilatation of the main and right pulmonary arteries. The left pulmonary artery (LPA) could not be seen. Angiography failed to show a LPA. This case of an absent LPA associated with absent pulmonary valve syndrome is discussed and the literature is reviewed. (Ann Thorac Cardiovasc Surg 2006; 12: 368–72)

Key words: absent left pulmonary artery, absent pulmonary valve

Introduction

Absent pulmonary valve syndrome is an uncommon form of congenital heart disease. It occurs in 2.4 to 6.3% of patients with tetralogy of Fallot (TOF).^{1,2)} The main symptoms are recurrent wheezes and dyspnea due to compression of the trachea and bronchi by the aneurysmal pulmonary arteries. This syndrome was first reported in 1830.^{3,4)} Calder et al.⁴⁾ analysed 245 patients with this syndrome, and found 17 cases of TOF with absent pulmonary valve associated with absent “origin” of the left pulmonary artery (LPA). They established that blood to its distal part was probably supplied through a patent ductus arteriosus in some and through pulmonary collateral channels in the others. They also reviewed 12 cases of the syndrome with anomalous origin of the LPA from the ascending aorta. The true (complete) absence of a main pulmonary artery (PA) branch was not critically reviewed. McCaughan et al.⁵⁾ reported a 14.3% incidence of absent

LPA in a group of 35 patients with absent pulmonary valve syndrome. This incidence of absent LPA is much higher than in patients with classical TOF (1.9%).⁶⁾

In this report a patients with this combination is discussed and compared to 15 similar cases reported in the literature.

Case Report

A six-year-old boy was referred to King Faisal Specialist Hospital and Research Center for cardiac evaluation. He was a result of a normal pregnancy and normal delivery. He had been well until one year of age when he started to develop mild cyanosis and dyspnea on exertion. On physical examination, the patient had mild cyanosis and clubbing of the fingers. There was no respiratory distress at rest. His weight was 15.6 kg (just below the 5th percentile for his age). Heart rate was 104 beats/minute and blood pressure was 100/55 mmHg. Cardiovascular examination revealed normal peripheral pulses, left parasternal heave, and to-and-fro murmur at the middle left sternal border. The second heart sound was single. Chest examination was normal, and there was no hepatomegaly. The electrocardiogram showed sinus rhythm, right axis deviation and right ventricular (RV) hypertrophy. Chest x-ray showed mild cardiomegaly, moderate prominence of

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Fig. 1. Pulmonary artery (PA) angiogram demonstrating absence of the left pulmonary artery (LPA) and aneurysmal dilatation of the right pulmonary artery (RPA). The left lung is oligemic.

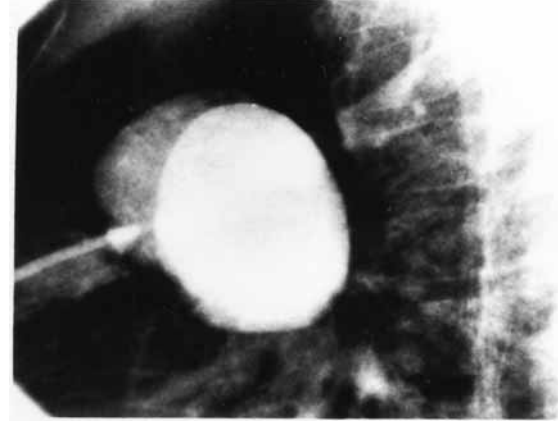


Fig. 2. Pulmonary artery (PA) angiogram demonstrating aneurysmal dilatation of the main and right pulmonary arteries, and pulmonary regurgitation.

the main PA and a small left lung. A 2-dimensional echocardiography and Doppler study confirmed the diagnoses of TOF and absent pulmonary valve. There was severe pulmonary regurgitation and a peak systolic gradient of 70 mmHg across the RV outflow tract (RVOT). There was severe dilatation of the main PA and right pulmonary artery (RPA); the RPA diameter was 29 mm. Accurate assessment of the LPA was hindered by the severe dilatation of the main PA and RPA. The aortic arch was left sided and no patent ductus arteriosus was seen.

At cardiac catheterization, RV systolic pressure was at systemic level and there was a peak systolic gradient of 77 mmHg across RVOT. The main PA pressure was 23/10 mmHg. The pulmonary flow to systemic flow (Q_p/Q_s) was 0.83. The catheter could not enter the site of LPA. PA angiography failed to show a LPA (Fig. 1) but clearly showed severe aneurysmal dilatation of the main PA and RPA and moderate pulmonary regurgitation (Fig. 2). Angiography following pulmonary vein wedge injection in the left lung failed to show LPA, confirming its absence. Aortography failed to show a patent ductus arteriosus, collaterals supplying the distal segment of LPA or anomalous origin of the LPA. The diagnosis of TOF, absent pulmonary valve and absent LPA was established.

Operative findings revealed large mal-alignment-type ventricular septal defect (VSD), overriding aorta, infundibular obstruction with hypertrophic muscle bundles, hypoplastic pulmonary valve annulus and absent pulmonary valve leaflets. The LPA was absent while the RPA was very dilated. The VSD was closed with a patch of bovine pericardium and the hypertrophic muscle bundles

in the infundibular area were resected. Rudimentary granular tissue, present in the area of the pulmonary valve, was excised and a transannular patch of bovine pericardium was used to enlarge the RVOT. The RV pressure was 50% of the systemic pressure at the end of the procedure. The patient came off the cardiopulmonary bypass successfully. He was extubated on the first postoperative day and discharged home 6 days later. On follow-up, it was noted that his symptoms had improved significantly. However, 3 years after surgery, his chest x-ray showed moderate cardiomegaly and echocardiography showed dilated RV with impaired function, dilated RPA (5 cm in diameter) and free pulmonary regurgitation. Cardiac catheterization revealed RV systolic pressure of 36 mmHg, end-diastolic pressure of 5 mmHg and RPA pressure 24/4 mmHg (mean 12). Four months later, the patient had a valved pulmonary homograft placement to avoid further impairment of RV function; the postoperative course was uneventful, and the patient was asymptomatic 5 years later.

Discussion

The combination of absent pulmonary valve syndrome and absent LPA is extremely rare. The literature was reviewed with regards to the occurrence of unilateral absence of a PA in patients with this syndrome. Only cases of “complete” unilateral absence of a PA (absence of both proximal and distal segments) were included. Cases in which the proximal segment of the PA branch was missing, with the distal segment being supplied through a ductus arteriosus or collateral, and cases of severe hypo-

Table 1. Data on patients with unilateral absence of pulmonary artery associated with absent pulmonary valve

Authors/ref. no.	Age at which symptoms started	Hemodynamics				The absent PA branch	Diagnosis confirmed by			Courses
		RV pr	PA pr	RV-PA pr	Qp/Qs		Angio	Surgery	Necropsy	
Nadas et al. ⁷⁾	6 h			NA		LPA	+	+	+	Died on 2 d postop., 1 mo old (extracardiac palliation failed)
Dixon et al. ⁸⁾	2 d	80	32	48	NA	LPA	+	+	NA	Died 1 y postop., 11 y old
Nagao et al. ⁶⁾	NA			NA		LPA	+	NA	NA	NA
Macartney and Miller ⁹⁾	NA (symptomatic)	80	25	55	1.7	LPA	+	NA	NA	Alive, 13 y old
Pinsky et al. ¹⁰⁾	NA (symptomatic)	90	25	65	1.4	LPA	+	+	NA	Died 6 mo postop., 5 y old
Arensman et al. ¹¹⁾	9 h	NA	NA	70	1.7	LPA	+	ND	NA	Died unoperated upon (infant)
	4 d	NA	NA	40	3.3	LPA	+	+	NA	Died 4 d postop., 5 mo old
	1 d	NA	NA	58	1.8	LPA	+	+	NA	Died 1 d postop., 13 y old
Presbitero et al. ¹²⁾	3 mo	100	28	72	0.3	LPA	+	+	+	Died 10 d postop., 9 y old
	1 w	130	50	80	2.5	LPA	+	ND	ND	Still alive, 43 y old
McCaughan et al. ⁵⁾	NA	NA	NA	*	NA	LPA	+	+		NA
	NA	NA	NA	*	NA	LPA	+	+		NA
	NA	NA	NA	*	NA	LPA	+	+		NA
	NA	NA	NA	*	NA	LPA	+	+		NA
	NA	NA	NA	*	NA	LPA	+	+		NA
Our case	1 y	100	23	77	0.83	LPA	+	+	-	Still alive, 4 y postop.

RV, right ventricle; pr, pressure; PA, pulmonary artery; RV-PA, peak systolic gradient across RV outflow tract; Qp/Qs, ratio of pulmonary to systemic blood flow; LPA, left pulmonary artery; NA, not available; ND, not done; postop., postoperative; h, hour; d, day; mo, month; w, week; y, year; *, present but severity is unknown.

plasia of the PA branch were excluded. Fifteen cases with absent LPA⁵⁻¹²⁾ were found and the patients in this report was added to them (Table 1). A true absence of RPA in association with absent pulmonary valve syndrome could not be found.

With respect to clinical features, all patients in whom data could be obtained were symptomatic with respiratory difficulty and or cyanosis and the symptoms started in the first week of life in 6 of them^{7,8,11,12)} and at 3 months of age in one.¹²⁾ The patient in this report was symptomatic by 1 year of age. The diagnosis of absent LPA was (or could have been) suspected from plain chest x-ray film in 10 patients,^{5,7-9,12)} as it showed hypoplasia of the left lung and significantly reduced vascularity.

Cardiac catheterization and angiography confirmed the absence of LPA and the marked dilatation of RPA in all patients. There was a gradient across the RVOT in 14 of them^{5,8-12)} and it ranged from 40 to 80 mmHg; in the remaining 2 patients,^{6,7)} this was not addressed. The direction of the shunt was stated only in 8 patients. It was left to right in 6 of them,⁹⁻¹²⁾ Qp/Qs ranged from 1.4 to 3.3. The shunt was right to left in the remaining 2. PA systolic

pressure was mentioned in 11 patients and it did not exceed 52 mmHg in any of them (McCaughan et al.⁵⁾ gave a range of 18 to 52 mmHg for the whole group including those with normal PA branches). There was only one patient with documented significantly elevated systolic PA pressure (50 mmHg) in whom cardiac catheterization was done at 38 years of age and she was still alive at 43 years of age.¹²⁾

Surgery was performed in 12 cases; intracardiac repair was performed in 11 of them. The result of total correction was documented in 6 of the 11 patients; there were 5 deaths,^{8,10-12)} 3 died within 30 days of the surgery,^{11,12)} 2 died at home 1 year and 6 months after the surgery (suddenly).^{8,10)} The present patient is still alive 5 years postoperatively. One female patient was still alive at 43 years of age without having had any surgery.¹²⁾ One infant died prior to surgery.

The main symptoms are usually recurrent wheezes and difficulty breathing due to external compression by the aneurysmal pulmonary arteries on the trachea and bronchi. This aneurysmal dilatation most likely occurs as a result of increased RV stroke volume. This is caused by

severe pulmonary regurgitation associated with absence of the pulmonary valve. In the absence of a main PA branch, the aneurysmal dilatation of the remaining PA may be exaggerated as the whole output from the RV is directed to it, and such patients are more symptomatic than those with normally developed RPA and LPA. Most of the patients reviewed were symptomatic.

Surgical correction of absent pulmonary valve syndrome in general is challenging and controversial. It has been met with high mortality especially when performed in infants who are significantly symptomatic, 58.3%.⁵⁾ Calder et al.⁴⁾ in their review of the literature demonstrated a 13.3–40% mortality following intracardiac repair in all age groups, with a higher mortality in infants. In the last decade the operative mortality has decreased to 11%.¹³⁾ In minimally symptomatic patients surgery can be delayed and intracardiac repair has a relatively low mortality rate, 5.0%.⁵⁾ The surgical correction of absent pulmonary valve when associated with absent main PA branch is even more challenging and probably carries a higher mortality rate; at least 5 of the 11 patients were dead within 1 year following the intracardiac repair, 3 of the deaths occurred within 30 days following surgery. It should be noted that in 4 of the deaths the age ranged from 5 to 13 years.

Although total correction to one lung without placement of a valve in the RVOT is possible, the long-term outlook is still unclear. It has been noted that pulmonary regurgitation following correction of absent pulmonary valve and TOF is well tolerated.^{5,10)} However, some patients may develop RV dysfunction secondary to long standing pulmonary regurgitation.¹⁴⁾ One factor that may enhance this complication in the unilateral absence of a PA is the development of pulmonary hypertension. Pool et al.¹⁵⁾ demonstrated a 19% incidence of pulmonary hypertension in unilateral absence of a PA in patients without a shunt and 88% incidence in patients with a shunt. Therefore, developing pulmonary hypertension may worsen pulmonary regurgitation and RV dysfunction. Initially in patients with minimal symptoms, insertion of a pulmonary valve may not be necessary but close follow-up is essential as valve insertion may be needed should RV dysfunction develop before this becomes irreversible. Another factor that should be assessed when considering the need for pulmonary valve insertion is the degree and course of the aneurysmal dilatation of the main PA branch as this can be progressive and severe enough to cause significant compression on the trachea and bronchi. A valve insertion (with total correction) cures pulmonary

regurgitation, which is the main stimulus for the continued pulmonary dilatation. It may at least ameliorate the dilatation of the proximal PAs and improve the symptoms. McCaughan et al.⁵⁾ recommended a valve insertion in all patients with this syndrome who have associated absence of one PA. The durability of a valved conduit in the RVOT is limited and has its own complications.¹⁶⁾ Homograft replacement of the central PA has been recommended for young infants with this syndrome when associated with severe respiratory distress as it improves survival.¹³⁾

Fortunately, the incidence of unilateral absence of a PA in patients with absent pulmonary valve syndrome is not high. Only 16 cases are documented in the literature, including our case. Patients with absence of “origin” of a main PA branch must be differentiated from those with true (complete) unilateral absence of PA as the management is different. Angiography following pulmonary vein wedge injection and aortography may be needed to exclude an abnormally developed (but present) PA such as an anomalous origin.

It has been suggested that the absence of a PA results from involution of the proximal 6th aortic arch (equivalent to extrapulmonary segment of the PA) in utero followed by regression of the fetal ductus arteriosus after birth leading to severe hypoplasia of the related intrapulmonary segment of the PA.¹⁷⁾

In conclusion, absent LPA is rarely associated with absent pulmonary valve syndrome and this combination carries higher morbidity and mortality. Although a valve insertion could be avoided during intracardiac repair in minimally symptomatic patients, it may eventually be needed because of RV dysfunction and therefore, close follow-up is crucial.

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