

## Absent Pulmonary Valve Associated with Tetralogy of Fallot and Complete Atrioventricular Septal Defect: Report of a Case

Seiya Kikuchi, MD,<sup>1</sup> and Masato Yokozawa, MD<sup>2</sup>

**A male infant with an extremely rare combination of absent pulmonary valve, tetralogy of Fallot and atrioventricular septal defect presented without symptoms of respiratory distress or congestive heart failure. He underwent successful primary repair at the age of 5 months. The procedure consisted of double-patch repair of the atrioventricular septal defect and right ventricular outflow tract reconstruction with a monocusp transannular patch. Resection or plication of a dilated pulmonary artery was not required. The patient is doing well without any symptoms 5 years after repair. (Ann Thorac Cardiovasc Surg 2005; 11: 44–7)**

**Key words:** absent pulmonary valve, tetralogy of Fallot, complete atrioventricular septal defect

### Introduction

Absent pulmonary valve is a rare congenital anomaly, characterized by dysplastic or absent pulmonary valve tissue and severe pulmonary regurgitation. Rarely isolated, it is usually associated with tetralogy of Fallot. The combination of absent pulmonary valve, tetralogy of Fallot and complete atrioventricular septal defect is extremely rare. There has been only one report of this combination in the English literature.<sup>1)</sup> We report a successful repair of this rare anomaly in an infant.

### Case Report

A newborn, full-term male infant was transferred to our institution because of mild cyanosis. There were no symptoms of respiratory distress or heart failure. Auscultation revealed a systolic murmur associated with an early diastolic murmur along the mid-left sternal border. The electrocardiogram showed right ventricular hypertrophy and incomplete right bundle branch block. The chest roent-

genogram demonstrated cardiomegaly with a cardiothoracic ratio of 61% and normal pulmonary vascular markings. Two-dimensional echocardiography demonstrated valvular pulmonary stenosis with pulmonary regurgitation, inlet type of ventricular septal defect with anterior extension, a large aorta overriding the ventricular septal defect (Fig. 1), a common atrioventricular valve with free-floating common anterior leaflet, primum atrial septal defect (Fig. 2), and right aortic arch. At 2 months of age, cardiac catheterization and angiography confirmed the diagnosis of tetralogy of Fallot, complete atrioventricular septal defect (Rastelli type C), absent pulmonary valve and right aortic arch. There was a 70 mmHg systolic pressure gradient across the pulmonary valve annulus with a pulmonary artery pressure of 15/9 mmHg. A pulmonary arteriogram (Fig. 3) demonstrated dilated pulmonary arteries, a distally displaced stenotic pulmonary valve annulus without pulmonary valve structure. A right ventriculogram (Fig. 4) demonstrated dilatation of the branch pulmonary arteries, and a left ventriculogram showed a gooseneck deformity of the left ventricular outflow tract and a right aortic arch.

Corrective surgery through a median sternotomy was performed when he was 5 months old. The right ventricular outflow was extremely elongated and extended to just below the bifurcation of the pulmonary artery. Thus the main pulmonary artery was almost absent and the pulmonary valve annulus was located just proximal to the

*From Departments of <sup>1</sup>Cardiovascular Surgery and <sup>2</sup>Pediatric Cardiology, Hokkaido Children's Hospital and Medical Center, Hokkaido, Japan*

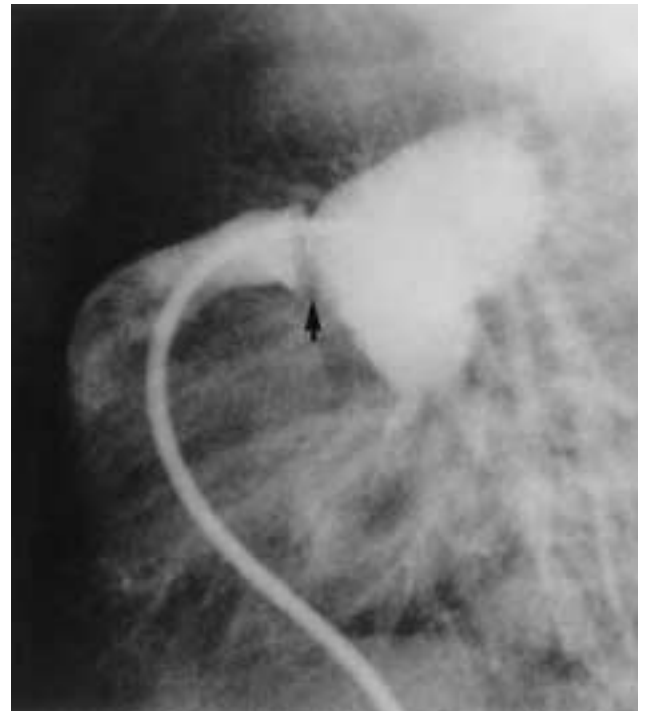
Received June 24, 2004; accepted for publication August 3, 2004. Address reprint requests to Seiya Kikuchi, MD: Department of Cardiovascular Surgery, Hokkaido Children's Hospital and Medical Center, 1-10-1 Zenibako, Otaru, Hokkaido 047-0261, Japan.



**Fig. 1.** Two-dimensional echocardiogram. Parasternal long-axis view showing a large aortic root overriding a ventricular septal defect with an enlarged right pulmonary artery. AO, aorta; RV, right ventricle; LV, left ventricle; LA, left atrium; RPA, right pulmonary artery.



**Fig. 2.** Apical four-chamber view showing characteristic features of type C complete atrioventricular defect. The free-floating common anterior leaflet rides over the crest of the ventricular septum and separates the primum atrial septal defect from the inlet ventricular septal defect. RV, right ventricle; LV, left ventricle; RA, right atrium; LA, left atrium



**Fig. 3.** Lateral view of pulmonary arteriogram demonstrating dilated pulmonary arteries and stenotic pulmonary valve annulus (arrow).

pulmonary artery bifurcation. Cardiopulmonary bypass was established with bicaval cannulation, and the heart was arrested with cold crystalloid cardioplegia. The pulmonary artery was longitudinally incised and then the incision was extended to the right ventricular outflow tract. Vestigial remnants of the leaflet of the pulmonary valve

were present at a small ventriculoarterial junction (Fig. 5). The complete atrioventricular septal defect was identified through a right atriotomy. Double-patch closure of the septal defects was performed. The mitral cleft was not closed. Right ventricular outflow obstruction was relieved with a monocusp transannular patch. The postop-



**Fig. 4.** Anteroposterior view of right ventriculogram demonstrating dilated pulmonary arteries, elongated right ventricular outflow tract and opacification of the left ventricle and aorta through a ventricular septal defect.



**Fig. 5.** Intraoperative photograph demonstrating vestigial remnants of the leaflet of the pulmonary valve (arrow).

erative course was uneventful. At 10 months of age, he underwent repeat cardiac catheterization to assess the hemodynamic effects of surgery. The peak systolic pressure gradient across the right ventricular outflow tract was 6 mmHg and there was no residual left to right shunting at ventricular level. Echocardiography performed at the

end of the follow-up period showed moderate pulmonary regurgitation, no mitral regurgitation, and mild tricuspid regurgitation. He is doing well without any symptoms 5 years following repair.

## Discussion

Since Chevers<sup>2)</sup> first described congenital absence of the pulmonary valve cusps, many cases of absent pulmonary valve syndrome have been reported. The lesion is rare in isolation and is more often associated with various heart defects, usually tetralogy of Fallot. A combination of absent pulmonary valve, tetralogy of Fallot and complete atrioventricular septal defect is extremely rare. To our knowledge, there has been only one English literature report of this unusual combination.<sup>1)</sup> Symptoms of absent pulmonary valve vary from life-threatening respiratory obstruction by aneurysmally dilated pulmonary arteries or cardiac failure in a neonate to absence of symptoms in patients who may live unrestricted lives for many years.

The markedly symptomatic neonate or infant should undergo a corrective operation involving insertion of a valve or valved conduit in the right ventricular outflow tract and partial resection, plication, or both of the aneurysmal pulmonary arteries.<sup>3-6)</sup> In contrast, the minimally symptomatic or asymptomatic patient, such as ours, should undergo a corrective operation electively in childhood. The insertion of a pulmonary valve prosthesis, and resection or plication of the aneurysmal pulmonary arteries is not essential.<sup>3)</sup>

## References

1. Vobecky SJ, Williams WG, Trusler GA, et al. Survival analysis of infants under age 18 months presenting with tetralogy of Fallot. *Ann Thorac Surg* 1993; **56**: 944–50.
2. Chevers N. Recherches sur les maladies de l'artère pulmonaire. *Arch Gen Med* 1847; **15**: 488–508.
3. McCaughan BC, Danielson GK, Driscoll DJ, McGoon DC. Tetralogy of Fallot with absent pulmonary valve: early and late results of surgical treatment. *J Thorac Cardiovasc Surg* 1985; **89**: 280–7.
4. Karl TR, Musumeci F, de Leval M, Pincott JR, Taylor JFN, Stark J. Surgical treatment of absent pulmonary valve syndrome. *J Thorac Cardiovasc Surg* 1986; **91**: 590–7.
5. Danilowicz D, Presti S, Colvin SB, Doyle EF. Repair in infancy of tetralogy of Fallot with absence of the leaflets of the pulmonary valve (absent pulmonary valve syndrome) using a valved pulmonary artery homograft. *Cardiol Young* 1992; **2**: 25–9.
6. McDonnell BE, Raff GW, Gaynor JW, et al. Outcome after repair of tetralogy of Fallot with absent pulmonary valve. *Ann Thorac Surg* 1999; **67**: 1391–6.