

# PULMONARY ATRESIA WITH RESTRICTIVE VENTRICULAR SEPTAL DEFECT: A RARE CONGENITAL HEART DISEASE

Abdullah A.A. Alabdulgader, MRCP; Omar Galal, PhD; Zoltan Halees, MD

Pulmonary atresia is a rare cyanotic congenital heart disease that may present with or without ventricular septal defect (VSD). When present with VSD, it is usually a non-restrictive membranous or infundibular defect.<sup>1</sup> Only two autopsy reports exist describing pulmonary atresia with restrictive VSD.<sup>2</sup> Our report presents the data of a patient with pulmonary atresia and a restrictive VSD, which has been confirmed echocardiographically and by cardiac catheterization. The patient underwent successful surgical repair.

## Case Report

A three-month-old female infant (weight 5.3 kg, length 58 cm) presented with central cyanosis that had been present since the first day of life. On physical examination, she showed severe cyanosis with mild respiratory distress, but with no dysmorphic features. Oxygen saturation measured by pulse oximetry ranged between 50%-60% in room air temperature. There was no edema. Abdominal examination revealed no organomegaly, and precordial palpation revealed no thrill—no heave and no palpable heart sounds. On auscultation, there were normal first heart sound, single second heart sound of soft pansystolic murmur grade 2/6 at the left sternal border, and third intercostal space with no significance radiation. There was no continuous murmur.

ECG showed a rate of 140beats/min. There was a right axis deviation with right ventricular hypertrophy. Chest radiograph showed levocardia, with stomach bubble on the left side. Echocardiogram revealed situs solitus-atrioventricular concordance, ventriculoarterial concordance and 4-mm perimembranous VSD, expanding a little to the outlet septum. It showed right-to-left shunt with Doppler gradient of 36 mm Hg. There was left aortic

From the Department of Pediatrics (Dr. Alabdulgader), King Fahad Hospital, Hofuf, and the Cardiovascular Department (Drs. Galal and Al Halees), King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia.

Address reprint requests and correspondence to Dr. Alabdulgader:

Ministry of Health, P.O. Box 9596, Hofuf 31982, Saudi Arabia.

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FIGURE 1. Right ventricular (RV) angiogram in four-chamber view. The RV cavity is well seen with the balloon catheter in the apex. There is right-to-left shunt through the small ventricular septal defect (VSD). The aorta arises from left ventricle (LV).

arch and small disconfluent pulmonary arteries with tiny patent ductus arteriosus.

Cardiac catheterization was performed. Access to the heart was obtained percutaneously through the right femoral vein and the left femoral artery. A size 4 French sheath was used on both sides. After having obtained pressures and saturations in the right and left sides of the heart (Table 1), angiography was performed in the right ventricle, aorta and pulmonary venous wedge in the left and right lung (Figure 1 and 2).

Angiography into the right ventricle revealed a good size right ventricle with mildly impaired contractility. There was no antegrade flow through the outflow of the right ventricle. A perimembranous VSD was demonstrated and it was very restrictive and measured about 4 mm in diameter. The VSD was shunting right to left opacifying the LV, and the ascending aorta was entirely arising from the left ventricle and not overriding as in the usual case of pulmonary atresia with VSD. There was no opacification of the pulmonary arteries from this injection (Figure 1). Thus, the final diagnoses were: a) situs solitus, levocardia of the left aortic arch; b) pulmonary atresia; c) restrictive (perimembranous) VSD; and disconnected pulmonary

FIGURE 2A. Left pulmonary venous wedge angiogram in the anterior posterior view showing retrograde filling of a hypoplastic left pulmonary artery

FIGURE 2B. Right-sided pulmonary venous wedge angiogram in the anterior posterior view showing retrograde filling of hypoplastic right pulmonary artery which ends well before the midline confirming discontinuity between pulmonary arteries.

TABLE 1. Hemodynamic data obtained at cardiac catheterization.

	Pressures	Saturations %
RA	4/1 mean is 3 mmHg	54
RV	93/6 mmHg	44
LV	64/6 mmHg	59
Aorta	82/56 mean 67 mmHg	62
IVC		53
SVC		51

Right ventricular pressure and left ventricular were obtained simultaneously but the aortic pressure was done at a different time. RA=right atrium; RV=right ventricle; LV=left ventricle; IVC=inferior vena cava; SVC=superior vena cava.

arteries supplied by a very small restrictive PDA. After establishing the diagnosis, the child underwent right ventricle-to-pulmonary artery confluent graft using 6 mm Gore-Tex tube. The VSD was left open as it was thought that the right ventricle would not tolerate the closure of the VSD. The patient did reasonably well after the procedure,

although she continued to be cyanosed. Echocardiogram done at this stage showed good forward blood flow through the right ventricular outflow tract, but there was evidence of stenosis at the origin of left pulmonary artery with pressure gradient of 35 mm Hg.

The patient was seen again in the clinic at the age of six months. She was found to have cyanosis with hemoglobin concentration of 12.6 g m/d L. ECG showed sinus rhythm rate of 110 beats/min, with right axis deviation and severe right ventricular hypertrophy.

At the age of eight months, the patient underwent a second cardiac catheterization. The body weight was 6 kg. The catheterization showed severe bilateral branch pulmonary arteries stenosis at the anastomosis of the conduit to the pulmonary arteries, and aortopulmonary collateral from descending aorta to the right pulmonary artery. She underwent a second surgery in the form of total correction by VSD closure using patch of bovine pericardium, and connection of the right ventricle to the pulmonary arteries with fresh pulmonary homograft (#14). She had a smooth postoperative course. Follow-up echocardiogram at the age of 14 months showed left pulmonary artery stenosis with pressure gradient of 50 mm Hg. There was also a gradient of 25 mm Hg where the homograft joins the right ventricle. Quantitative perfusion scan was done at this stage. This showed a flow to the left lung of 23% and the to the right lung of 77% of the total cardiac output. In view of these results, the patient was taken to the cardiac catheterization laboratory where a stent was placed at the origin of the left pulmonary artery. This resulted in disappearance of the gradient. Balloon dilatation was done for the right ventricular out flow tract stenosis with reasonable result.

## Discussion

Pulmonary atresia with ventricular septal defect is a rare cyanotic congenital heart disease. Our report presents an infant with pulmonary atresia and a restrictive VSD which was managed by a two-stage surgery. At the age of 15 months, our patient was stable at home. Unlike our case, the two previously reported cases died at surgery and the diagnosis was established by cardiac catheterization and at autopsy.<sup>2</sup> The first case simulated pulmonary atresia with intact ventricular septum presenting at three months of age with venous congestion, severe tricuspid regurgitation and suprasystemic right ventricular pressure. Because of increasing cyanosis, the patient underwent subclavian

artery-to-pulmonary artery anastomosis with atrial septectomy, but died during the procedure. Autopsy showed a large VSD obstructed by tricuspid valve tissue. The second case had right ventricular systolic pressure that was initially (at 13 months) at the systemic level but had increased to suprasystemic level by three years of age. The patient improved after a left subclavian artery to pulmonary

artery anastomosis. At five years, he was restudied in anticipation for intracardiac repair. He died during attempted construction of right ventricular outflow tract using valved conduit. Autopsy showed large VSD obstructed by hypertrophied septal band.<sup>2</sup>

In two reviews dealing with the clinical, hemodynamic and surgical aspects of pulmonary atresia and VSD in 50 and 80 patients, respectively,<sup>3,4</sup> no mention was made of obstruction of VSD. A third extensive review of the pathological anatomy in 172 cases of pulmonary atresia with VSD (106 of which were uncomplicated or simple) similarly makes no mention of obstruction of the VSD.<sup>5</sup> Musewe et al.<sup>6</sup> reported 80 new cases of tetralogy of Fallot over a four-year period. Of these, five patients were identified as having restrictive VSD during routine echocardiography. Four of the five patients had hemodynamic, angiographic and surgical confirmation of restrictive VSD, with additional necropsy confirmation in one. The mode of VSD restriction in the four patients was felt to be tricuspid septal leaflet or accessory tags.

Looking at the embryological closure of the interventricular foramen shows that the mechanism of VSD closure depends on three factors: continued growth of connective tissue on the crest of the muscular septum; downward growth of ridges dividing conus and truncus arteriosus; and projections into the arterioventricular canal from the right-sided cushions.<sup>7</sup> By these explanations, some authors have postulated that restrictions of ventricular septal defect are due to accessory tricuspid valve tissue, anomalous insertion of accessory tissue into the margin of the ventricular septum, or the hypertrophied septal band.<sup>2,6</sup> Surgical findings in our case revealed the tricuspid valve shrouded over the ventricular septal defect, indicating that the most likely mechanism in our case was accessory tricuspid valve tissue.

We conclude that pulmonary atresia with restrictive VSD can be diagnosed echocardiographically and angiographically. According to the size of the pulmonary artery, different surgical approaches are needed. In our case due to hypoplastic pulmonary arteries, a two-stage repair was found useful.

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