SUNTEXT REVIEWS

SunText Review of Case Reports & Images
ISSN: 2766-4589

Open Access
Case Report
Volume 2:1

Complete Atrioventricular Septal Defect with Absent Pulmonary Valve in an Infant with Down syndrome: A Rare Association

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Received date: 14 June 2021; **Accepted date:** 21 June 2021; **Published date:** 28 June 2021

Citation: Yildırim AI, Karaagac AT, Usta SA, Erolu E (2021). Complete Atrioventricular Septal Defect with Absent Pulmonary Valve in an Infant with Down syndrome: A Rare Association. SunText Rev Case Rep Image 2(1): 120.

DOI: https://doi.org/10.51737/2766-4589.2021.020

Abstract

Absent pulmonary valve syndrome (APVS), defined as the absence or hypoplasia of the pulmonary valve, stenosis of the pulmonary valve annulus, and aneurysmal dilatation of the pulmonary trunk and its branches, is rare congenital heart disease with an incidence of 0.2%-0.4%.

Keywords: Atrioventricular Septal Defect; Pulmonary valve syndrome

Introduction

Absent pulmonary valve syndrome (APVS), defined as the absence or hypoplasia of the pulmonary valve, stenosis of the pulmonary valve annulus, and aneurysmal dilatation of the pulmonary trunk and its branches, is rare congenital heart disease with an incidence of 0.2%-0.4% [1,2]. It is commonly classified into two types: The first type shares many similarities with tetralogy of Fallot (TOF), including a ventricular septal defect, overriding aorta, and absent ductus arteriosus, so is called TOF-type APVS. The other less common type with an intact ventricular septum, a mild degree of pulmonary artery dilatation, and a patent ductus arteriosus is also called non-TOF-type APVS [3]. This congenital anomaly may be associated with other cardiac pathologies, but its association with atrioventricular septal defect (AVSD) is extremely rare. Apart from heart complications, APVS has a poor prognosis mostly due to the aneurysmal dilatation of the pulmonary trunk and its branches, which can compress the bronchus and esophagus, leading to nutritional and respiratory complications [3,4].

Case Report

A 3-month-old full-term female infant with Down syndrome was accepted to our hospital with the signs of respiratory infection. She had been hospitalized several times with signs of respiratory distress before being accepted to our hospital. On her physical examination, she had tachypnea, tachycardia, bilateral crepitant and subcrepitant rales, 3/6 systolic murmur along the mid-left sternal border on auscultation, and a 3-4 cm palpable liver below the right lower costal margin. Her laboratory findings were normal except for moderately increased acute phase reactants and mild anemia. The electrocardiogram showed left axis deviation and sinus tachycardia. Cardiomegaly and prominent pulmonary vascular bed were observed on her chest roentgenogram. The transthoracic echocardiography revealed unbalanced complete atrioventricular septal defect (AVSD), left ventricular hypoplasia, secundum type atrial septal defect (ASD), absent pulmonary valve, and pulmonary arterial aneurysm (Figure 1).

On her computed thorax tomography, pulmonary annulus was 8 mm and bifurcation was observed. The pulmonary arteries were enlarged, the right pulmonary artery was 23 mm and the left one was 16 mm in diameter, thus applying compression on both right and left main bronchi. Besides, bilateral subsegmental atelectasis

and mosaic perfusion pattern were observed due to the effect of pressure (Figures 2 and 3).

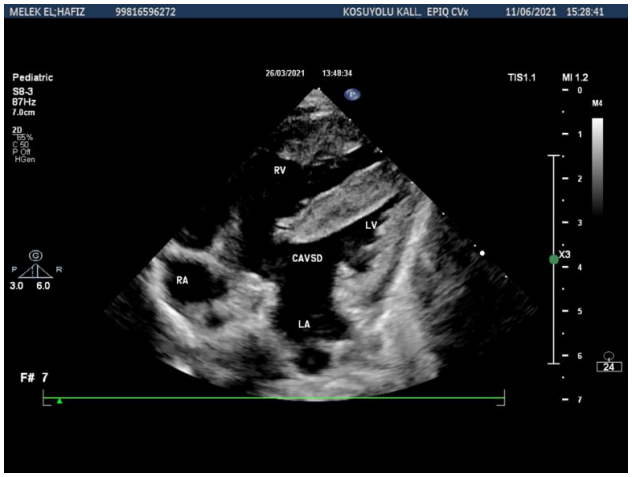

Figure 1: *Echocardiographic imaging of atrioventricular septal defect, left ventricular hypoplasia, atrial septal defect, and absent pulmonary valve.*

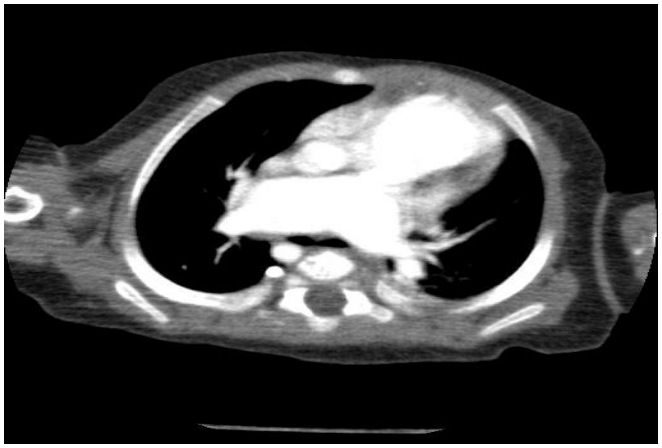

Figure 2: *Torax CT image showing the compression of enlarged pulmonary arteries on the pulmonary bronchi.*

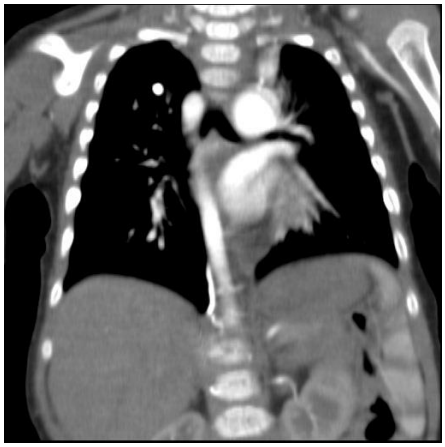

Figure 3: *Thorax CT image showing the effect of compression on pulmonary parenchyma.*

Dilatation was detected in the proximal esophagus as a result of the left pulmonary arterial compression. A transcatheter angiography was performed after her respiratory infection was treated. Angiography confirmed the absence of pulmonary valve leaflets and revealed a 90 mmHg gradient across the pulmonary valve annulus with a mean pulmonary arterial pressure of 19 mmHg.

Our patient, who was a candidate for a single ventricle operation due to the unbalanced AVSD, underwent pulmonary banding and surgical plication as the first stage operation. She didn't experience any post-operative problems and was discharged with the planning of the Fontan procedure following a Glenn operation at the most convenient time for her. She has been followed successfully for 5 months.

Discussion

APV is defined as a syndrome because it is associated with cardiac (tricuspid atresia-intact septum, absent aortic valve-intact septum, Ebstein's anomaly, tetralogy of Fallot, interrupted aortic arc), extracardiac (CHARGE association, agenesis of corpus callosum, duodenal atresia, bilateral renal agenesis) or chromosomal (trisomy 13, trisomy 18, trisomy 21, 22q11 deletion) anomalies [5,6]. Respiratory distress and infection are common in these patients due to the compression of the bronchial tree by aneurysmatic dilatation of the pulmonary arteries [7]. Our patient had tachypnea, tachycardia, bilateral crepitant and subcrepitant rales, 3/6 systolic murmur along the mid-left sternal border on auscultation, and a 3-4 cm palpable liver below the right lower costal margin on admission. She had trisomy 21 and AVSD, a very rare association with the absent pulmonary valve, free pulmonary valve insufficiency, and accompanying severe annular artery aneurysm on cardiologic evaluation. Two main types of APVS have been described in the literature as TOF type, associated with the ventricular septal defect, overriding aorta, and absent ductus arteriosus, and non-TOF type [3]. Our case differed from these two classifications because it was accompanied by an unbalanced AVSD. The anatomic data required for pre-operative evaluation of patients with atrioventricular septal defect and/or absent pulmonary valve syndrome may be confidentially obtained by transthoracic echocardiography [5]. In our case, unbalanced complete atrioventricular septal defect, left ventricular hypoplasia, secundum type atrial septal defect, absent pulmonary valve, and pulmonary arterial aneurysm were highly suggestive of absent pulmonary valve syndrome, but the rarity of the association prompted cardiac catheterization. Angiocardiography confirmed the absence of pulmonary leaflets and the mean pulmonary arterial pressure was measured before surgery. The prognosis of APVS is poor mainly due to the extra cardiac complications resulting from compression of pulmonary bronchi and esophagus. In addition to medical treatment of cardiac and

respiratory pathologies, various methods for pilication of the absent pulmonary valve, surgical reduction of a pulmonary aneurysm, and pulmonary valve repair are applied for the surgical management [4-10]. In the study of Young et al. early mortality rate was reported as 13,5% and 5-10-20 year average late mortality rate as 6,7% in the 52 postoperative APVS patients [11]. In the present case, due to the presence of unbalanced AVSD, pulmonary banding and pilication operation was performed initially. The patient was discharged in good health. Glenn and Fontan surgeries will be planned in the most suitable time during her follow-up.

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