Isolated absent pulmonary valve with intact ventricular septum in a young child: A rare case report

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ABSTRACT

Absent pulmonary valve syndrome, commonly linked with tetralogy of Fallot and ventricular septal defect, is a rare congenital condition. It is exceedingly rare to have an isolated absent pulmonary valve with an intact ventricular septum without cardiovascular shunt lesions, such as an atrial/ventricular septal defect or patent ductus arteriosus. This report presents a case of such rarity involving a young child with recurrent lower respiratory tract infections.

Keywords: Absent pulmonary valve, congenital heart disease, intact ventricular septum

INTRODUCTION

Isolated absent pulmonary valve with an intact ventricular septum in the absence of cardiovascular shunt lesions such as atrial/ventricular septal defect or patent ductus arteriosus is exceedingly rare. This report presents a case of such rarity in a young child.

CASE REPORT

A 2-year-old acyanotic boy visited the pediatric outpatient clinic due to recurring lower respiratory tract infections. Clinical evaluation showed cardiomegaly and a low-pitched early diastolic murmur in the pulmonary area. A chest radiograph showed cardiomegaly with a prominent right ventricular outflow tract (RVOT) [Figure 1]. Echocardiography revealed dilated right-sided heart chambers, dilated tricuspid valve and pulmonary annulus, and absent pulmonary valve leaflets with free pulmonary regurgitation. No other structural cardiac defects were seen. Subsequently, the patient underwent a computed tomography angiography (CTA) to delineate the intracardiac anatomy. CTA revealed an absent pulmonary valve with a dilated right atrium, right ventricle, and RVOT with intact interatrial and

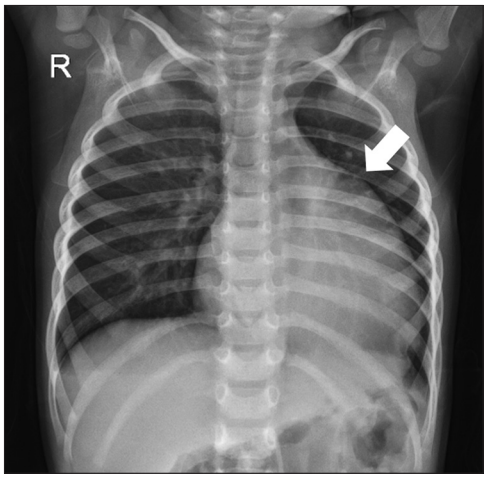

Figure 1: Chest radiograph showing cardiomegaly with a prominent right ventricular outflow tract (white arrow)

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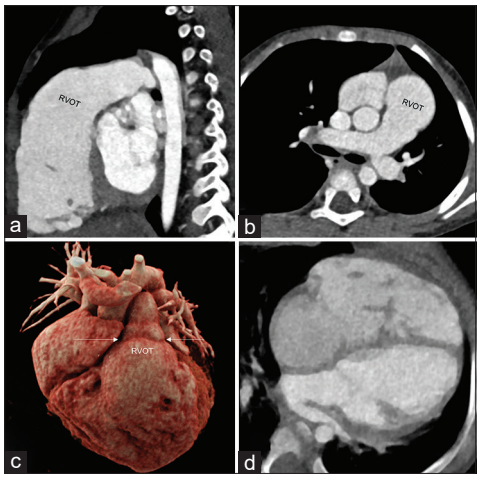

Figure 2: Computed tomography angiography (CTA) images (a and b) showing dilated, right ventricle, right ventricular outflow tract (RVOT), and no visible pulmonary leaflets. Cinematic rendered CT image (c) demonstrating the dilated RVOT with the region of the pulmonary annulus (white arrows). Four-chamber CTA image (d) shows intact interventricular and intact interatrial septum. RVOT: Right ventricular outflow tract

interventricular septum [Figure 2]. The main, right, and left pulmonary arteries were of normal size. No other cardiovascular anomalies were detected, thus leading to a diagnosis of an isolated absent pulmonary valve. The child was currently advised of medical follow-up with an elective surgery planned in few years.

DISCUSSION

Absent pulmonary valve syndrome (APVS) was initially described by Cheevers *et al.* in 1847.[1] Typically, APVS is associated with atrial or ventricular septal defects, patent ductus arteriosus (PDA), or tetralogy of Fallot. The occurrence of APVS with an intact ventricular septum is infrequent, with limited cases reported in the literature. Previous cases of absent pulmonary valve with intact ventricular septum have consistently been found to have PDA.[2-4] Nagar, Deendayal, *et al.*[5] reported a case of an 18-year-old girl who presented with progressive dyspnea on exertion and was found to have an absent pulmonary valve with severe pulmonary regurgitation and a small atrial septal defect. She had an intact ventricular septum with absent PDA. Attie *et al.*[6] documented an isolated absent pulmonary valve in a 66-year-old man without any intracardiac shunt lesions. Okajima *et al.*[7] published a case report of a 79-year-old woman who presented with worsening right heart failure. On evaluation, she was found to have APVS with IVS without any other associated cardiovascular shunt lesion.

To the best of our knowledge, the present case represents a rare instance of an isolated absent pulmonary valve with an intact ventricular septum without associated cardiovascular anomalies in a young child. This case underscores the importance of CTA in defining intricate anatomy and identifying potential associated cardiovascular anomalies in patients suspected of having APVS.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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