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Absent Pulmonary Valve Associated With Tetralogy of Fallot and Double Chamber Right Ventricle

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Introduction: Tetralogy of Fallot (TOF) in the absent of pulmonary valve is a rare congenital anomaly (3%of TOF patients).
Case Presentation: We are presenting an 18- year-old male with history of dyspnea on exertion and cyanosis since childhood. Despite the diagnosis of congenital heart disease, no intervention was done due to his poor socioeconomic culture. At age of 18, he referred to our center for further evaluation of heart problem. After physical examination and other diagnostic tests, our final diagnosis was tetralogy of fallot, absent pulmonary valve and double-chamber right ventricle. The absence of mature PV tissue leads to severe pulmonary regurgitation, which is often associated with significant dilatation of the pulmonary arteries. After further evaluation of main pulmonary artery, its branches and other abnormalities of the heart, total correction of tetralogy and pulmonary valve replacement was done. His post operation period was uneventful without any complication and he discharged after 10 days.
Conclusions: We believe that the prognosis for those patients in this era is good and complete surgical treatment is tolerated well by them.

Keywords: Pulmonary Valve; Tetralogy of Fallot; Double Chamber Right Ventricle

1. Introduction

Tetralogy of Fallot (TOF) with absent pulmonary valve (PV) is an uncommon congenital anomaly characterized by features of TOF with either undeveloped ridges or the total absence of pulmonic valve tissue. Congenital absence of the PV with an intact ventricular septum may occur, but this is greatly less common. The absence of mature PV tissue leads to severe pulmonary regurgitation with significant dilatation of the pulmonary arteries, which is the characteristic of this syndrome. An interesting aspect of this anomaly is that the ductus arteriosus is often absent (1). TOF is the most frequent cause of cyanotic congenital heart disease and may occur at a rate of 1-3 cases per 1000 live births. However, TOF with absent PV is rare, with about 3% of patients with TF having the absent PV syndrome (1). The present case is an interesting case with absent PV, TOF and fascinatingly associated double-chamber right ventricle (DCRV).

2. Case Presentation

An eighteen year old male was born in Iraq. He was noted to be cyanosed at birth. His parents knew he had congenital heart disease but due to war and poor economy, they could not offer to do surgery. During childhood, he had dyspnea on minimal exertion. By the age of 15, he had adopted a lifestyle that avoided any physical exertion. Finally, he referred for further evaluation and surgery. On admission, he had dyspnea on exertion (NYHA functional class III). Physical examination revealed BP = 110/70mmHg, HR = 75bpm and oxygen saturation = 86% in room air, elevated jugular vein pressure, normal S1, single S2, load III/VI systolic murmur on upper left sternal boarder with III/VI decrescendo diastolic murmur. Also he had clubbed extremities and peripheral cyanosis.

His laboratory data was included: Hemoglobolin = 16.9 mg/dl, Mean cell volume (MCV): 87 micro m3, Ferritin = 210 micro g/litr, Platelet count: 158000/micro litr, Uric acid = 5.6 mg/dl, Creatinine: 0.8 mg/dl; Indeed this patient had mild iron deficiency, and thrombocytopenia. His Electrocardiogram showed normal sinus rhythm, heart rate = 75bpm, right axis deviation, RA abnormality, tall R in all precordial leads, RV hypertrophy and right bundle branch block (RBBB). Figure 1 shows his chest X-ray with normal to decreased lung vascularity, RV enlargement and prominent pulmonary artery.

Implication for health policy/practice/research/medical education:
This case report demonstrated a rare case of absent pulmonary valve without lung problem in adulthood. We believe these findings will be of interest to the readers of your Journal.

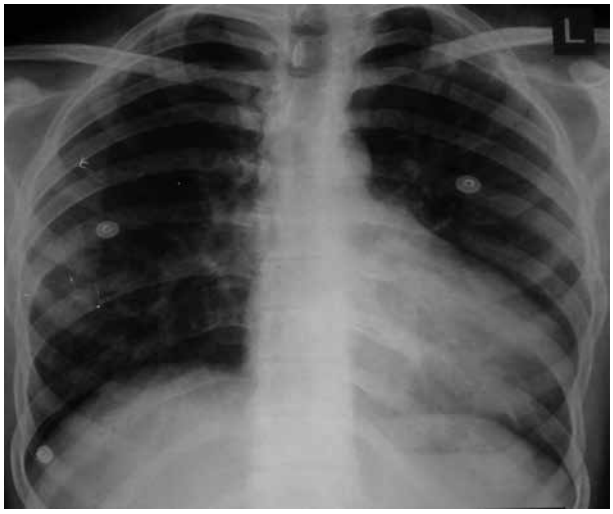

Figure 1. Normal Chest Wall, No Lung Disease, Normal to Decreased Lung Vascularity, RV Enlargement, Prominent Pulmonary Artery

Transthoracic echocardiography (TTE) revealed abdominal and atrial situs solitus, levocardia, ventricular looping was D-loop with normal relation of great arteries; the superior and inferior vena cava drained normally into the right atrium. He had normal left ventricular size with preserved function, moderate to severe right ventricular (RV) enlargement, severe RV hypertrophy associated with anomalous muscle band suggestive for double-chamber RV and mild to moderate RV dysfunction, large size ventricular septal defect, overriding of aorta and interestingly absent PV resulting in severe pulmonary regurgitation, dilated main pulmonary artery and its branches specially right pulmonary artery (Figure 2, 3). So echocardiography with color flow imaging and Doppler interrogation establishes the diagnosis and hemodynamic consequences of TOF with absent PV. Computed tomography-Angiogram (CT-angio) was done for further evaluation of peripheral pulmonary vasculature and lung parenchymal which revealed normal arborization of pulmonary arteries, No major aorto-pulmonary collateral arteries (MAPCA) (Figure 4). Finally, our patient underwent catheterization. Coronary angiography showed normal coronary arteries, DCRV with 90 mmHg pressure gradient, Normal pulmonary artery pressure, Free pulmonary regurgitation, Dilated pulmonary arteries; with the following pressures data: LV: 130/0-12 mmHg, Aorta: 130/80 mmHg, RV: 100/0-4 mmHg (low RVEDP is probably due to low preload and also his RV function was mild- moderately reduced), Pulmonary artery: 18/4 mmHg; and saturation study of aorta showed Ao saturation of 86%. So our final diagnosis was TOF with absent PV, severe free pulmonary regurgitation and DCRV. The patient underwent total correction of TOF and removal of hypertrophied RV muscle bundle and PV replacement. He had good post operation period without any complications. Post operation TTE showed small residual VSD, hemodynamically normal PV without paravalvular leakage. Patient discharged without any complication.

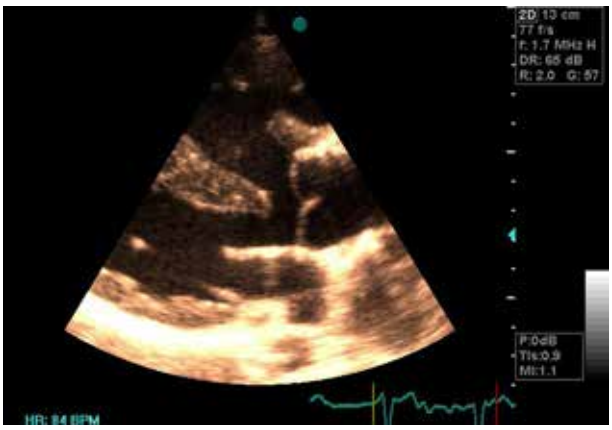

Figure 2. TTE in Parasternal Long Axis View Showing Over-ridded Aorta and Large Sub-aortic Ventricular Septal Defect

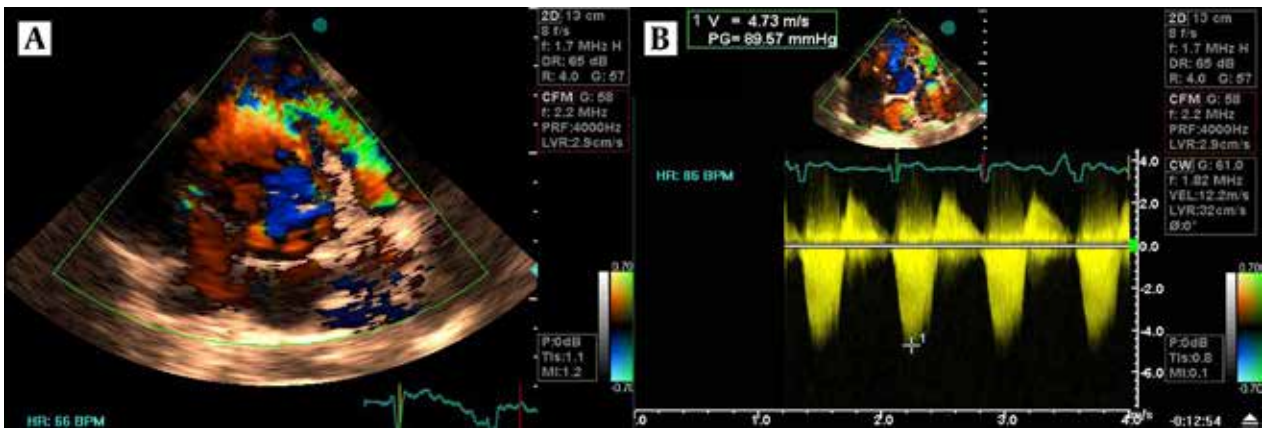

Figure 3. A) Severe Obstruction in Mid Portion of the Right Ventricle; B) Severe Pulmonary Regurgitation,

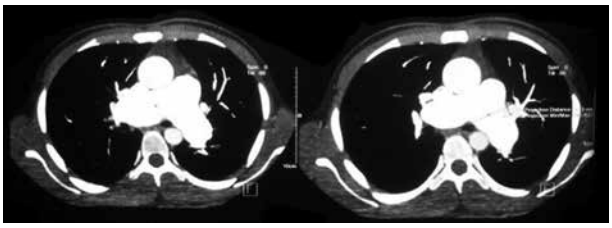

Figure 4. Normal Arborization of Pulmonary Arteries Associated with Dilated PA

3. Conclusions

The first report of the association of absent PV and ventricular septal defect was done by Royer and Wilson in 1908 (2). Their case was studied at autopsy and unexampled because the patient also had corrected transposition of the great arteries in situs inversus (L-transposition in situs inversus). Absent PV syndrome is a rare congenital cardiac malformation (1, 3). Most frequently, the intra-cardiac anatomy is TOF (4), but the characteristic morphologic feature of this lesion is either the complete absence of the PV leaflets or the presence of remnants that are usually dysplastic nodules guarding a small ventriculo-pulmonary junction. This feature results in some degree of RV outflow tract obstruction, significant pulmonary regurgitation, RV enlargement, and often significant dilation of pulmonary arteries (4, 5). Although a male predominance has been commented on this syndrome, it is not confirmed by the reported cases. Of 116 reported cases the sex was indicated in 77, and in those the male: female ratio was about 1:1 (37 male: 40 female) (6). Although our reported patient is in adulthood period, the majority of patients with TOF and absent PV present in infancy or early childhood with heart failure, cyanosis or airway obstruction due to the compression from aneurysmally dilated main PA or PA branches (7). On physical examination of these patients, a pulmonary ejection sound is absent despite dilation of pulmonary trunk because PV is absent (8); also in our case, the pulmonary component of the second heart sound is absent because there is no PV (8). Often the aortic component of second heart sound is muffled by interposition of dilated pulmonary trunk (9). A midsystolic murmur is maximum in the second left intercostals space and is loud, harsh, and long because a large RV stroke volume is ejected across a narrow annulus into a dilated pulmonary trunk (9, 10). The regurgitant diastolic murmur is usually grade 3/6, short and harsh, ending well before subsequent first heart sound. The combination of a long, harsh, loud systolic murmur followed by a shorter harsh diastolic murmur creates the characteristic auscultatory feeling of sawing wood (11). Similar to our patient on ECG exam, the P wave is frequently both peaked and tall. The QRS axis is rightward. When the PV is absent, the tall monophasic R wave in lead V1 extends into adjacent precordial leads in contrast to TOF in which the tall right precordial R wave is characteristically limited to lead V1 (12). On CXR of these patients, often the pulmonary trunk and proximal branches dilate, massively. Dilated RV occupies the apex, and an enlarged right atrium forms the right lower cardiac silhouette. Pulmonary vascularity usually is normal than decreased (13). Indeed the pulmonary arterial tree shows several attractive features in these patients. The main and proximal portions of pulmonary arteries are usually dilated with frequent aneurysmal dilatation of one branch of the pulmonary artery, more commonly the right one (13); and about the other diagnostic tests, as mentioned above, the echocardiography is the main and non-invasive tool for comprehensive study and CT-angiography and catheterization can help in evaluation of details, completion of study and decision making for management. The prognosis of patients with absent PV and ventricular septal defect is variable (12, 13). The long term outcome of these patients has been evaluated in several series with survival rates from 82-93% at the first year and 79-87% at five years (14, 15). There are few reported cases in adulthood. The oldest one was 51 year old lady with respiratory symptoms for years, which had good postop period and early resolution of symptoms and signs of RV failure (16). We believe that the prognosis for those patients in this era is good and complete surgical treatment is tolerated well by them. Our patient is unique regarding to associated DCRV and absent PV syndrome and successful corrected surgery in adulthood.

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Authors' Contribution

All authors have contributed in this case report.

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