


Aortopulmonary window in adults: A rare entity leading to Eisenmenger syndrome

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Abstract

An aortopulmonary window (APW) is a rare congenital heart defect involving an abnormal communication between the ascending aorta and the pulmonary trunk with separate aortic and pulmonary valves. This defect accounts for 0.2% of all congenital cardiac anomalies and if left untreated can lead to Eisenmenger syndrome, severe pulmonary hypertension, heart failure, and poor survival. The authors herein present a case of APW type III with Eisenmenger syndrome in an adult patient whose initial complaint was cyanosis, and provide a thorough review of the literature of cases of APW with Eisenmenger syndrome that have survived into adulthood.

KEYWORDS

2D echocardiography, cardiac imaging, color Doppler, congenital heart defects, congenital heart disease

1 | INTRODUCTION

The aortopulmonary window is an anatomical communication between the ascending aorta and the pulmonary trunk in the presence of two separate aortic and pulmonary valves creating a left-to-right shunt. Aortopulmonary window (APW) is a rare form of congenital heart disease that accounts for 0.2% of all congenital heart defects.¹ The survival of patients with untreated large APW defects is very poor, with 40% mortality in the first year of life.^{2,3} If corrective surgery is initially delayed, severe pulmonary hypertension, Eisenmenger syndrome, as well as congestive heart failure develop in the first few months of life.⁴ Once irreversible pulmonary vascular disease develops, especially when the shunt reverses and becomes right-to-left, corrective surgery is no longer an option.⁵ Late presentation of APW in adult life is infrequent and, hence, there is limited literature on the presentation and management of APW beyond early childhood.⁶ With APW being a rare type of congenital heart defect with very poor survival if not surgically corrected, very few untreated cases are expected to survive into adulthood.

1.1 | Objectives

The objective of this review was to report on all cases present in the literature of adult patients with aortopulmonary window and Eisenmenger. We also describe the case of a 24-year-old man with APW type III and Eisenmenger syndrome who presented to the outpatient department of a tertiary care medical center with the chief complaint of cyanosis.

2 | LITERATURE REVIEW AND METHODS

A literature review of the published cases of APW with Eisenmenger syndrome in the adult population was conducted using the following databases: MEDLINE (1946–2018), PubMed, and Google Scholar. Also, references of retrieved papers were checked for other relevant references. The terms included: “aortopulmonary window or aortopulmonary septal defect”, and “eisenmenger or eisenmenger syndrome or eisenmenger complex”. The search was limited to adults and yielded 8 published papers reporting survival of patients with

TABLE 1 Summary of reported cases of APW with Eisenmenger syndrome in adults

Case report	Gender and age at presentation/ death	Imaging modality of diagnosis	Symptomatic or asymptomatic	Type/location of APW	Year of publication	Other
Sharma et al ¹⁷	33-y-old Female	2D TTE showed large defect between ascending aorta and main pulmonary trunk 3D TTE showed APW is proximal to semi-lunar valves CT further diagnosed interrupted aortic arch	Symptomatic since childhood Bluish discoloration of body, thickening of nail beds, dyspnea on exertion, and repeated respiratory infections in childhood and infancy	APW Type 3 with interrupted aortic arch and Eisenmenger	2015	
Bobylev et al ¹⁴	32-y-old Male	CT/MRI diagnosed APW TTE did not diagnose APW but showed a structurally normal heart, dilated LV and RV, and global hypokinesia EF of 20%	Symptomatic since childhood Dyspnea and cyanosis with progressive cardiopulmonary decompensation	APW with Eisenmenger (type not mentioned)	2014	Heart and lung transplant were done and the patient survived
Su-Mei et al ¹⁸	Died at 60 y of age Female	2D TTE showed large APW measuring 3.2 cm with severe RVH and PHTN	Was undiagnosed for over 40 y of age Lived relatively asymptomatic until her 50s then developed exertional dyspnea and deteriorated gradually	APW with Eisenmenger (type not mentioned)	2007	Died from asystolic cardiac arrest Longest surviving documented case of APW Gave birth to 3 children in her thirties
Nadig et al ¹⁹	18-y-old Male	2D TTE diagnosed APW Contrast echocardiography confirmed the presence of APW with right-to-left shunt 64 slice CT further confirmed the diagnosis	Symptomatic Presented with easy fatigability, exertional dyspnea, gum bleeding and intermittent headaches in the past year	APW Type 1 with Eisenmenger	2014	Presented with polycythemia

(Continues)

TABLE 1 (Continued)

Case report	Gender and age at presentation/ death	Imaging modality of diagnosis	Symptomatic or asymptomatic	Type/location of APW	Year of publication	Other
Hari Krishan Aggarwal et al ²⁰	25-y-old Female	CECT chest showed communication between ascending aorta and pulmonary trunk CT angiography confirmed APW TTE did not diagnose APW, but showed PHTN and dilated RA and RV	Symptomatic Occasional episodes of cyanosis and palpitations on exertion during last 3 y	APW just above aortic and pulmonary origin, with Eisenmenger (type not specified)	2015	Successfully completed first pregnancy
Kose et al ²¹	27-y-old Female	Right heart catheterization with aortic root injection diagnosed APW TEE and TTE did not diagnose APW but showed dilated RA, RV, and TR	Asymptomatic up to first pregnancy at age 27. Severe dyspnea 3rd day postoperatively of C-section No respiratory symptoms until 7th mo of pregnancy, then developed exertional dyspnea	APW with Eisenmenger (type not specified)	2015	
Niles et al ²²	Died at 46 y of age Female	Cardiac catheterization diagnosed APW	Symptomatic since childhood. Chest pain, recurrent URTI, and increasing fatigability, cyanosis and cough	APW with Eisenmenger (type not specified)	1980	Pregnancy was terminated early to avoid complications Autopsy of heart showed that the common supravulvular chamber measured 8 cm in lateral and anteroposterior diameters and 12 cm vertically
Balegadde et al ²³	32-y-old Female	Diagnosed by multidetector CT with CT pulmonary angiography of the chest Cardiac MRI further diagnosed APW type 1 TTE did not diagnose APW, but showed dilated RA, RV, and PA	Asymptomatic until 31 y of age. Presented with major complaints of heart failure (class II–III) ongoing for 4 mo No presyncope/syncope or cyanotic spells	APW Type 1 with Eisenmenger (developed at age 33)	2018	PHTN 125 mm Hg

Abbreviations: CECT = contrast enhanced computed tomography; C-section = cesarean section; CT = computed tomography; EF = ejection fraction; MRI = magnetic resonance imaging; PA = pulmonary artery; PHTN = pulmonary hypertension; RA = right atrium; RV = right ventricle; RVH = right ventricular hypertrophy; TEE = transesophageal echocardiography; TR = tricuspid regurgitation; TTE = transthoracic echocardiogram; URTI = upper respiratory tract infections.



FIGURE 1 2D and 2D color Doppler echocardiographic view showing a large communication between the aorta and the pulmonary artery representing aortopulmonary window type III. 2D = two-dimensional; Ao = aorta; PA = pulmonary artery

APW and Eisenmenger into adulthood, describing a total of 9 cases. The study selection was accomplished through two steps. First, the literature was reviewed as a first step and then all case reports that describe patients of interest were included. Afterward, full articles were obtained for all accepted studies. Excluded articles included those that do not discuss APW with Eisenmenger syndrome in an adult or articles in any language other than English.

Table 1 summarizes the published cases found in the literature review according to gender, age at presentation or age at death, imaging modality of diagnosis, whether or not patient was symptomatic at presentation with reported symptoms, type or location of the APW, year of publication, and other pertinent and relevant information. The table serves as a quick reference for the currently available literature.

3 | CASE

A 24-year-old man presented to our outpatient clinic reporting episodes of central and peripheral cyanosis, dyspnea upon exertion, and palpitations that had started around the age of 9 years. At that time, he had sought medical attention at a peripheral hospital in Syria where he was diagnosed with a congenital heart defect with Eisenmenger syndrome via cardiac catheterization. However, upon presentation to our clinic, the patient had no clear report documenting the anatomy of his genetic defect nor was he aware of the nature of his congenital heart defect. Furthermore, on physical examination, patient was found to have jugular venous distention, bilateral upper and lower extremity clubbing, and pectus carinatum. Upon auscultation, patient had a holosystolic murmur in the tricuspid area, a prominent P2, and parasternal left heave exaggerated with inspiration. His blood pressure was 101/49 mm Hg, heart rate 51 beats/min, and oxygen saturation 91 percent at rest that dropped to 79 percent with walking. Laboratories were drawn and showed a hemoglobin level of 155 g/L, MCV of 71 fl, and a ferritin level of 2.25 pmol/L (normal 56.2–629.2 pmol/L). The patient was started on oral iron therapy, and pulmonary arterial hypertension therapies were discussed.

EKG findings at presentation showed right ventricular hypertrophy. Transthoracic echocardiography was done and showed levocardia and atrial situs solitus with the finding of a large 39 mm isolated

aortopulmonary window type III with Eisenmenger syndrome (Figure 1). The color Doppler study demonstrated trace physiological tricuspid and pulmonary insufficiency, with right-to-left shunting at APW level with no gradient between the pulmonary artery and the aorta. This is indicative of an estimated systemic or even supra-systemic pulmonary artery pressure, keeping in mind that cardiac catheterization remains the gold standard in accurately assessing pulmonary artery pressures.

4 | DISCUSSION

Aortopulmonary window is one of the rarest congenital heart diseases resulting from a communication between the proximal aorta and the main pulmonary artery. Such an abnormality can be isolated, such as in the case of our patient; however, in 50 percent of patients, it can be associated with other cardiac abnormalities such as coarctation of the aorta, interrupted aortic arch, tetralogy of Fallot, and atrial septal defect.⁷ Early correction is essential to prevent irreversible pulmonary vascular disease. According to a classification system proposed by Mori et al,⁸ APW can be classified into 3 types, a proximal, distal, or a combined proximal and distal type. Type I, the proximal type, is when the communication is located above the pulmonary and aortic valves between the ascending aorta and pulmonary trunk. Type II, the distal type, is when the communication involves the pulmonary bifurcation at the level of the right pulmonary artery. Type III, the rarest type, which is present in our patient, is characterized by the complete absence of the aortopulmonary septum resulting from a combination of the proximal and distal defects (Figure 2).

To the best of our knowledge, there have been 8 reported cases of APW with Eisenmenger syndrome in an adult dating back to 1980 of which 6 out of the 8 cases were female. One patient had an APW type 3 defect, another 2 patients had an APW type 1 defect, and for the remaining patients, the type of APW was not specified. Age of patients ranged from 18 to 60 years old and 4 of the 8 reported cases were symptomatic since early childhood. Symptoms included exertional dyspnea, cyanosis, palpitations, respiratory infections, bleeding of gums, intermittent headaches, and easy fatigability. The other half of reported cases remained asymptomatic until the age of 22, 27, 31, and 50 years keeping in mind, however, that symptoms do

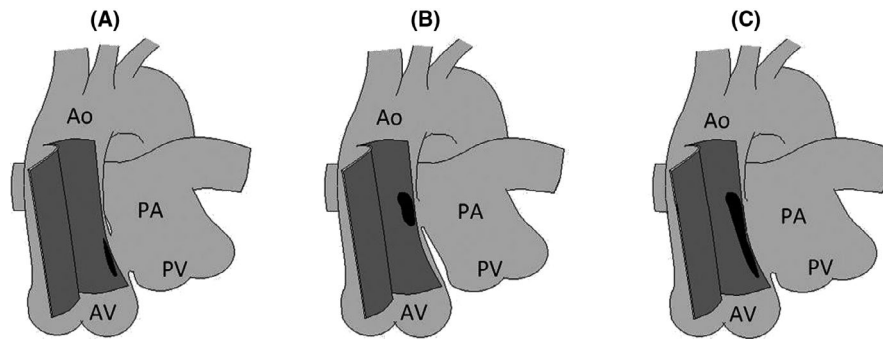


FIGURE 2 Black-shaded area representing (A) Type I, proximal defect, midway between the semilunar valves and the pulmonary bifurcation. B, Type II, distal defect, with the posterior border absent and aortic origin of the right pulmonary artery. C, Type III, total defect, incorporating defects present in both types of aortopulmonary window. Ao = aorta; AV = aortic valve; PA = pulmonary artery; PV = pulmonary valve

not necessarily correlate with the severity of the underlying disease. As for survival, one patient died at the age of 46 years and another died at the age of 60 years, the longest surviving documented case of APW (Table 1). Moreover, the literature search retrieved two reported cases of aortopulmonary window in symptomatic adult females aged 22 and 30 years old that had not developed Eisenmenger syndrome and where preoperative evaluation with right heart catheterization was in favor of surgical repair.^{9,10} In the case reported by Myers et al,⁹ the patient had significant pulmonary hypertension postoperatively that had improved at 13 months of follow-up. For one of the patients, the APW defect was type 1,⁹ whereas for the other, the type was unspecified.¹⁰

The general treatment strategy for patients with Eisenmenger is mainly based on clinical experience. Phlebotomy should be considered in patients with severe hyperviscosity symptoms and a hematocrit above 65%. Supplemental oxygen therapy has also been used for patients in whom it produces a consistent increase in arterial oxygen saturation, and anti-pulmonary hypertensives, which include prostanooids, endothelin receptor antagonists, and phosphodiesterase-5 inhibitors that target endothelial dysfunction, have been approved for the treatment of pulmonary arterial hypertension. Moreover, patients need to be counseled on general recommendations for physical activity, pregnancy, infections, air travel, exposure to high altitudes and elective surgery, and psychological support.¹¹

For the few cases of patients with APW who are not Eisenmenger but may have established pulmonary hypertension without reversal of the shunt, surgical repair is possible if certain strict criteria are satisfied. Generally, the operability of patients with pulmonary arterial hypertension associated with congenital heart disease is based on the likelihood of a favorable outcome vs an unfavorable outcome after repair resulting in biventricular circulation. Strict hemodynamic criteria, pulmonary vascular resistance (PVR), the ratio of PVR to systemic vascular resistance (SVR), and the change in their value in response to a vasodilator challenge have been suggested for the assessment of operability in these patients.¹² A baseline PVR index of <6 WUxm² and a resistance ratio <0.3 have been used as an indicator for a favorable outcome after corrective surgery leading to biventricular circulation, with no indication for vasoreactivity

testing. If the baseline PVR index is between 6 and 9 WUxm² with a ratio of PVR to SVR of around 0.3–0.5, then vasodilator challenge (with oxygen or nitric oxide) is strongly encouraged and it is broadly accepted that these patients are surgical candidates if all of the following criteria are met: a decrease of PVR index of 20%, PVR to SVR ratio decrease of 20%, final PVR index <6 WUxm², and a final PVR to SVR ratio <0.3 .¹² A heart–lung transplant can still be an option in such patients that are not eligible for repair.^{13,14} Some authors however question how accurately hemodynamic measures can determine risk of death or whether or not the patient will develop persistent PVR following repair as other factors such as the type of cardiac defect or genetic predisposition of the patient can alter hemodynamic testing or have an impact on outcome after surgical correction.^{15,16} Such concerns therefore create a lack of consensus as to whether vasoreactivity testing is accurate enough to determine whether patients will or will not have a good long-term outcome.

We therefore present a review of adult cases of APW with Eisenmenger syndrome in order to emphasize the importance of early surgical correction to avoid irreversible pulmonary hypertension.

CONFLICTS OF INTEREST

The authors have no conflict of interest to report.

AUTHOR CONTRIBUTIONS

Joud El Dick, and Mariam Arabi were the main authors of this article, performed the literature review, and wrote the manuscript. Mariam Arabi and Joud El Dick attended to the patient and followed up on his care in the outpatient department clinic and the Children's Heart Center at The American University of Beirut. Issam El-Rassi, Christelle Tayeh, and Fadi Bitar critically reviewed the manuscript and Issam El-Rassi made the drawing shown in Figure 2.

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