Case Report

A rare case of large aortopulmonary window with Eisenmenger syndrome and adult survival

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\textbf{ABSTRACT}

Patients with polycythemia have an abnormally elevated hemoglobin, hematocrit, and red cell count. It is important to differentiate primary polycythemia from secondary causes since this can affect patient management and prognosis. We report the case of a young male, suspected to have primary polycythemia who was referred for a cardiology opinion after a bone marrow examination was normal and testing for Janus kinase gene mutation was negative. Presence of central cyanosis and clubbing, clinical evidence of severe pulmonary artery hypertension, and significant hypoxemia on arterial blood gas analysis suggested that the polycythemia was secondary to an intracardiac shunt. Transthoracic and contrast echocardiography revealed a large aortopulmonary window with right-to-left shunting. A 64-slice cardiac computed tomography imaging confirmed the diagnosis. In the developing world, it is not uncommon to encounter such unusual cases; careful attention to basic clinical signs and use of multimodality imaging are helpful in establishing the correct diagnosis.

\textbf{Learning objective:} Clinicians should be aware that it is important to distinguish between primary and secondary polycythemia. Although Eisenmenger syndrome is rare in the West, it is not uncommon to encounter such cases in the developing world. Since such patients are polycythemic, they often undergo unnecessary hematological investigations as happened in our case. Careful attention to basic clinical signs and a systematic approach with multimodality imaging were helpful in establishing the diagnosis.

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Introduction

Polycythemia is usually defined as an increase in the hemoglobin levels above normal. Primary polycythemia or polycythemia vera (PV) is a genetically mediated myeloproliferative disorder while secondary polycythemia is seen in heavy smokers, chronic pulmonary disease, and cyanotic congenital heart disease. It is important to clinically distinguish patients with primary polycythemia from those with secondary causes. Presence of central cyanosis, clubbing, physical cardiac signs, and an abnormal arterial blood gas analysis are clues to the presence of an underlying intracardiac shunt as a cause of secondary polycythemia.

Although the incidence of Eisenmenger syndrome has dramatically declined in the West, it is not uncommon to encounter such cases in the developing world, since patients often remain undiagnosed and present late. An aortopulmonary (AP) window is a rare cause of Eisenmenger syndrome and results from an abnormal septation of the truncus arteriosus [1]. Most such defects present with early onset congestive heart failure during infancy and adult survival in surgically uncorrected patients is rare [2,3].

We report the case of a young male suspected to have primary polycythemia, referred for a cardiology opinion after a bone marrow examination was normal and testing for Janus kinase (JAK) gene mutation was negative. Careful clinical examination, 2D and contrast echocardiography followed by 64-slice computed tomography (CT) imaging confirmed the diagnosis of a large underlying AP window and Eisenmenger physiology causing secondary polycythemia.

Case report

An 18-year-old male presented with easy fatigability, exertional breathlessness, gum bleeding, and intermittent headaches since 1 year. There was no history of recurrent chest infections or hospitalizations in early childhood. His initial investigations revealed elevated hemoglobin (23 g/dl), red blood cell count of 7.37 million/cc, and a hematocrit of 70. Other biochemical
parameters were normal. Considering the possibility of primary polycythemia, the patient had undergone a bone marrow examination which revealed normoblastic erythroid hyperplasia. Testing for JAK gene mutation was negative. He was referred for a cardiology opinion to our institute.

The patient had a plethoric face with bilateral conjunctival suffusion; despite the ruddy complexion, central cyanosis and bilateral symmetrical clubbing were evident. There was no audible murmur; prominent pulmonary artery pulsations with loud P2 component of the second heart sound were noted suggesting severe pulmonary artery hypertension (PAH). At the time of the first medical examination, the measured SpO2 was 83%.

Arterial blood gas (ABG) analysis revealed resting oxygen desaturation (\(\text{SO}_2\) of 85%) with arterial hypoxemia (\(\text{PO}_2\) of 56 mmHg). The 12-lead electrocardiogram (ECG) revealed sinus rhythm, superior frontal QRS axis with biventricular hypertrophy (ECG on half voltage in precordial leads, Fig. 1). Mild cardiomegaly and a dilated main pulmonary artery segment were seen on the chest X-ray. Although an echocardiogram performed elsewhere had reported only severe PAH, in view of the clinical findings and hypoxemia on the ABG, we repeated the echocardiogram to look for an intracardiac shunt that had possibly resulted in secondary polycythemia.

There was no shunt at the atrial or ventricular level but a clear echo drop-out was visible between the aorta and pulmonary artery in the parasternal short-axis view (Fig. 2). A contrast echocardiography using agitated saline confirmed the absence of any right-to-left shunt at the atrial or ventricular level (Fig. 3; Video 1). The agitated saline was seen opacifying the aorta in the parasternal short-axis view, confirming the presence of an AP window with right to left shunting (Fig. 4; Video 2). A 64-slice cardiac CT with intraaortic endoscopic reconstruction was performed which delineated a huge Type I AP window (Fig. 5, panels A and B).

**Discussion**

It is important to differentiate primary from secondary polycythemia since this can affect patient management and prognosis. Important clinical clues to differentiate between them include presence of cyanosis, clubbing, and auscultatory evidence of severe PAH, as were evident in this case, leading to suspicion of underlying intracardiac shunt. Analysis of ABG is also important in
differentiating between primary and secondary polycythemia. In patients with primary polycythemia, arterial oxygen saturation is usually normal as compared to those with secondary causes [4,5]. However, occasionally even in primary polycythemia, mild desaturation may occur due to the hyperviscosity which prevents proper diffusion of oxygen in the alveolar capillary system leading to subtle pulmonary abnormalities [6]. Our case had mild arterial desaturation (85%); however, significant arterial hypoxemia (PO2 of 56 mmHg) was suggestive of an intracardiac shunt.

With early detection and treatment of congenital heart disease in the West, Eisenmenger syndrome is now rarely encountered. However, it is not uncommon in the developing world to encounter such cases, as patients often go undetected in early life and seek medical attention late. AP window is an uncommon congenital anomaly of conotruncal septation, occurring in about 0.15% of all congenital heart diseases [1]. The most common is Type I variety, due to a deficiency in proximal portion of the AP septum; Type II (distal) and Type III defects (with anomalous origin of the right pulmonary artery from the aorta) are rarer [7]. Our case belonged to the usual Type I variety.

In most cases of AP window, the defect is nonrestrictive leading to early congestive heart failure and failure to thrive in infancy. The prognosis of surgically uncorrected AP window is poor, with most patients dying during infancy or early childhood. Long-term adult survival with AP window with development of Eisenmenger syndrome, although uncommon, is known [2,3]. This case was unusual since the patient had undergone work-up for primary polycythemia, including a bone marrow examination and genetic studies. Presence of clubbing, the abnormal ABG analysis, and the echocardiographic findings helped in establishing the correct diagnosis.

Conclusion

Clinicians should be aware that it is often important to distinguish between primary and secondary polycythemia. In this case too, a normal bone marrow examination and negative testing for JAK mutation prompted a cardiology referral. Careful attention to basic clinical signs and a systematic approach with the aid of multimodality imaging were helpful in establishing the diagnosis.

Conflict of interest

The authors declare no conflict of interest.

Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at http://dx.doi.org/10.1016/j.jccase.2014.07.008.

References