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Platypnea-Orthodeoxia Syndrome With Atrial Septal Defect and Ectatic Aortic Root: A Case Report and Review of the Literature

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Abstract

Platypnea-orthodeoxia syndrome (POS) is a rare and underdiagnosed disease characterized by dyspnea in the upright position (platypnea) with simultaneous hypoxemia (orthodeoxia) that is relieved by recumbency. The physiopathological mechanisms involved are mediated by intracardiac shunts, pulmonary arteriovenous shunts or ventilation/perfusion mismatch. When POS is caused by a cardiac pathology, there is an anatomical (interatrial communication) and a functional component (as a dilated aorta or pneumectomy) working together to cause a right to left shunt without a constant right to left pressure gradient. Diagnosis is suspected through pulse oximetry verifying orthodeoxia. Confirmation usually is made by transesophageal echocardiography with bubble study to visualize the shunt. Percutaneous closure of the shunt is effective in most cases of cardiac POS. We report a case of an 87-year-old woman with POS related to a patent foramen ovale and an ectatic aorta followed by a review of the literature.

Keywords: Platypnea-orthodeoxia syndrome; Patent foramen ovale; Atrial septal defect; Aortic diseases

Introduction

Platypnea-orthodeoxia syndrome (POS) is a rare condition characterized by dyspnea in the upright position (platypnea) that is relieved by recumbency. Simultaneously, a dramatic decrease in arterial blood oxygen saturation in a sitting or standing position is easily observed and related to a simultaneous change in arterial blood gas sample (orthodeoxia) [1, 2].

Although pulmonary or hepatic diseases may be the causes of POS [1, 3, 4], the most common etiology is cardiac and related to an interatrial communication without constant right-to-left (R-L) pressure gradient but with an R-L shunt that occurs preferably in the upright position [5].

We present a case of POS associated with an occult patent foramen ovale (PFO) and an ectatic aorta followed by a review of the literature.

Case Report

An 87-year-old woman was admitted to our emergency medicine department complaining of severe dyspnea within the last 3 h. She denied cough, sputum production or fever. On physical examination, her blood pressure was 130/56 mm Hg, pulse was 79 beats per minute and respiration rate was 32 breaths per minute. Pulmonary auscultation was clear. Cardiac examination revealed a soft 2/6 systolic ejection murmur along the left sternal border. The oxygen saturation was 87% breathing at room air. Arterial blood gas analysis (ABG) at room air showed pH 7.55, pCO_2 23 mm Hg, pO_2 46 mm Hg. With a high flow mask, her oxygen saturation improved to 91%. Analytic study showed no relevant changes. Coagulation study was normal. Chest roentgenogram revealed an apparent dilated aorta with clear lung fields. Electrocardiogram was in sinus rhythm without any other electrical changes. Chest computed tomography (CT) and pulmonary CT angiography showed no parenchymal or vascular problem. She was admitted to our Internal Medicine Intermediate Care Unit for further evaluation.

Her past medical history was significant for a pulmonary embolism 3 years ago. At that time, the chest CT with angiography showed a central bilateral pulmonary embolism and the transthoracic echocardiography revealed right ventricular dilatation and dysfunction. She was submitted to thrombolysis with subsequent resolution of hypoxemia.

Some months after this event, she was admitted to our Internal Medicine Department due to pyelonephritis. During her stay, frequent periods of dyspnea and desaturation with poor response to oxygen therapy were noticed, which prompted further evaluation. Ventilation-perfusion scan did not show any perfusion amputation. Chest CT showed some bronchiectasis and fibrotic striatae in both pulmonary bases, without interstitial lung disease. Pulmonary function tests and electromyo-

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Table 1. ABG Collected With the Patient in Different Positions

	Supine position	Sitting position
PaO_2 (mm Hg)	83.5	56.4
Oxygen saturation (%)	98	88
O_2 concentration	0.5	0.5

graphy were normal. Transthoracic echocardiography showed an aneurysmatic interatrial septum without apparent interatrial communication, no pulmonary hypertension and normal biventricular function.

Chronic hypoxia was assumed to be probably due to previous pulmonary thromboembolism. She was discharged with domiciliary oxygen therapy and oriented to pneumology outpatient consult.

When we first saw the patient in our Intermediate Care Unit, we noticed the symptoms appeared to worsen on moving from supine to erect position. Pulse oximetry showed a decrease in the oxygen saturation to less than 80% in the upright position and an increase to more than 90% in the recumbent position.

This worsening was confirmed through ABGs in both positions, revealing marked orthostatic desaturation (Table 1). This pattern was consistent with platypnea-orthodeoxia.

Having the information of the previous two transthoracic echocardiograms, we decided that the best course of action was to first exclude an intracardiac shunt. Contrast-enhanced transthoracic echocardiography with agitated saline in the supine and erect position did not show an intracardiac shunt but indicated a possible extracardiac R-L shunt. This led us to make a new chest CT with angiography in a venous and an arterial phase to exclude intrapulmonary shunts. Again, the exam was negative for shunts but revealed an aneurysmatic aortic root (44 mm) already present in previous chest CTs.

At this point, we had a patient with POS, without liver disease, significant pulmonary disease or apparent intracardiac shunt.

Keeping in mind the aneurysmatic interatrial septum and a transthoracic echocardiogram with agitated saline suggesting the presence of a shunt, we decided to do a transesophageal echocardiography that showed an aneurysmatic interatrial septum with phasic protrusion to the left atria, associated to a PFO with a low magnitude left-to-right shunt. The inferior vena cava flux was turbulent and directed to the atrial septum. We could not confirm an inversion of the shunt due to poor collaboration of patients in a Valsalva maneuver.

We proceeded to a cardiac catheterization that showed normal pulmonary artery pressure (15/6 mm Hg) and normal right atrial pressure (5 mm Hg).

The patient's PFO was closed percutaneously using a 22 mm septal occluder device placed in the interatrial septum. A post-procedure echocardiographic bubble study was negative for shunting.

The patient's dyspnea immediately resolved. Her oxygen saturations on room air in the upright position improved to more than 94% and she was discharged completely asymptomatic.

Discussion

This syndrome was first reported in 1949 by Burchell in a case of intrathoracic arterial venous shunt [6]. Later, in 1969 and 1976, Altman and Robin respectively, used the terms platypnea-orthodeoxia to describe a syndrome, at this time, mostly in patients with hepatic or pulmonary diseases [7, 8]. It was only in 1984 that Seward and colleagues reported a series of patients with POS related with interatrial communications with R-L shunts without pulmonary hypertension [2].

There are three known causes for POS, namely, cardiac, pulmonary and hepatic ones. The physiopathological mechanisms involved are mediated by intracardiac shunts, pulmonary arteriovenous shunts and ventilation/perfusion mismatch [3, 4, 9].

When POS is caused by a cardiac disease, there is an anatomical and functional component working together to cause an R-L shunt without a constant R-L pressure gradient. So, anatomically, we find an interatrial communication as a PFO, atrial septal defect (ASD) or an atrial septal aneurysm with septal fenestration.

Because left atrium pressure is higher than right atrium pressure, there is no R-L shunt through a PFO or a small ASD. But, this kind of shunt can occur if we add a functional component that inverts the flow through the shunt.

A transient pressure elevation in the right atrium (hemodynamic explanation) induced by physiological maneuvers (posture change, inspiration, Valsalva maneuver, and cough) [10] and some diseases (right ventricular myocardial infarction, pulmonary embolism, constrictive pericarditis and pericardial effusion [11-14]) can provide this functional premise.

In the same way, some anatomical distortions can change the blood flow direction through the shunt (flow phenomenon) like in conditions such as emphysema, pneumonectomy, kyphoscoliosis and aortic aneurysm or elongation [1, 4, 14].

In the presence of this last condition, orthostatism could stretch the interatrial communication, augmenting the flow through the shunt or it could displace the atrial septum towards the horizontal position directing the blood flow from the inferior vena cava to the atrial septum, thereby extending the shunt [1, 9, 14, 15].

PFO is present in about 25-30% of healthy individuals [16] but is normally asymptomatic. In our patient, PFO was accompanied by an elongated, ectatic thoracic aorta and kyphoscoliosis. Because aortic root dilatation is a progressive and age-dependent process, this patient just developed symptoms of POS much latter in her life.

Several mechanisms have been proposed to explain how an ectatic aorta induces POS. Such an enlargement seems to be able to alter atrial septal geometry promoting a more direct pathway for the blood to flow from the vena cava to the interatrial communication or making the interatrial septum more mobile and permeable if there is an underlying defect [14, 15].

Even though this article is not focused on the non-cardiac causes of POS, we are going to make a brief reference to pulmonary arteriovenous shunts and ventilation/perfusion mismatch as physiopathological mechanisms involved in pulmonary and hepatic causes.

The presence of pulmonary arteriovenous shunts is anoth-

er mechanism behind POS and we can find it, for example, in patients with pulmonary arteriovenous malformations/fistulae. Blood passes through pulmonary arteriovenous shunts without being oxygenated in the lungs. When the patient is in the upright position, gravity increases blood flow in the lung bases. Usually there are a great number of shunts in the lower lung fields, so the upright posture will increase the arteriovenous shunt, with consequent POS [4, 9].

On the other hand, in the hepatopulmonary syndrome, intrapulmonary vascular dilatation (IPVD) is the structural abnormality behind reduced arterial oxygenation in the setting of liver disease. IPVD increases pulmonary blood flow without changes in alveolar ventilation inducing a ventilation-perfusion mismatch. At the same time, there is an increased passage of mixed venous blood, through intrapulmonary shunts, into the pulmonary veins. IPVD is more common in the lung bases that normally are already overperfused (zone three phenomenon), especially in the upright posture, resulting in an exacerbation of the ventilation-perfusion mismatch as well as in arteriovenous pulmonary shunt and consequent POS [17, 18].

POS is in fact a rare disease, but probably underestimated, and its diagnosis may be particularly difficult unless there is a high index of suspicion. The key to the diagnosis is a good clinical history and physical exam with relatively simple tests (comparison of pulse oximetry and arterial blood gas analyses performed with the patient in different positions) but most of the diagnostic procedures will be falsely negative when they are done in the supine position.

When an intracardiac R-L shunt is suspected, contrast-enhanced transthoracic echocardiography after injection of agitated normal saline in the supine and upright position is very sensitive, and allows definitive establishment of diagnosis in most cases [4, 19, 20]. In our patient, these exams were not conclusive and even misdirected our study to an intra-pulmonary shunt. This false negative result with saline contrast has been already described by some authors and is a relevant pitfall to account when trying to diagnose PFO [21]. A contrast tilt-table transesophageal echocardiography in lying and upright position may be needed. In some few cases, the shunt was seen only on Valsalva maneuvers [22, 23].

Percutaneous closure of the intra-atrial communication is very effective as the treatment of choice in most cases. After the procedure, most patients will have a complete resolution of their symptoms and will be able to resume a normal life [12, 24-26].

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