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Case Report

Platypnea-Orthodeoxia Syndrome: A Rare Cause of Dyspnea

Joana Freitas Ribeiro [1,*], Cátia Gorgulho [1], Débora Lopes [2], Ana Matos [1], Luís França [2], Nuno Catorze [2]

[1] Internal Medicine Service, Unidade Local de Saúde do Médio Tejo, Abrantes, Portugal.
[2] Intesive Medicine Service, Unidade Local de Saúde do Médio Tejo, Abrantes, Portugal.

* Correspondence: joanaffreitasribeiro@gmail.com.

Abstract: Platypnea-Orthodeoxia Syndrome (POS) is a rare and underdiagnosed condition characterized by dyspnea and hypoxia in the upright and sitting positions, which resolve in the supine position. The authors report the case of a 79-year-old woman with progressively worsening dyspnea over three days. In the emergency department, dyspnea and desaturation were observed, requiring increasing oxygen therapy when standing. Laboratory tests, electrocardiogram, and chest computed tomography revealed no abnormalities to explain the condition. Transthoracic and transesophageal echocardiograms identified a right-to-left shunt caused by a large atrial septal aneurysm, which was treated with closure using an Atriasept – CARDIA™ prosthesis. The procedure led to immediate symptom resolution. POS diagnosis is primarily clinical. While cardiac catheterization is the gold standard for diagnosis, echocardiography plays a crucial role in identifying the underlying etiology. Treatment depends on the cause, and this condition is associated with a good prognosis.

Keywords: Platypnea; Orthodeoxia; Dyspnea; Hypoxia; Shunt.

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1. Introduction

Platypnea-Orthodeoxia Syndrome (POS), first reported in 1949 by Burchell et al., is a rare condition requiring a high degree of suspicion and is likely underdiagnosed. It predominantly affects elderly individuals and women [1-6]. Currently, there are no data to estimate the incidence of POS; however, a 2017 literature review identified only 150 articles including a total of 239 patients [2]. Clinically, this syndrome is characterized by the onset of dyspnea and hypoxia in the upright or seated position, which resolve when lying down (platypnea – dyspnea triggered by the upright position; orthodeoxia – hypoxia in the vertical position) [1].

Although the pathophysiology of POS has puzzled the scientific community for years and, in some patients, the exact mechanisms remain unknown, three recognized etiologies exist: intracardiac shunts, pulmonary diseases with ventilation/perfusion mismatches, and hepatic diseases with pulmonary arteriovenous shunts. The most common etiology is cardiac defects, with a patent foramen ovale being the leading cause of intracardiac shunts, followed by atrial septal defects and aneurysms of the atrial septum [6]. POS arises when, in addition to these structural abnormalities, there is a functional component that promotes right-to-left shunting through the defect, such as the upright position.

The authors aim to raise awareness of POS, particularly its clinical presentation, diagnostic challenges, and therapeutic approaches.

2. Case Report

A 79-year-old woman with a medical history of hypertension, hypothyroidism, cerebrovascular disease, and osteoarticular disease presented to the emergency department with holocranial headache without alarm signs and dyspnea that had progressively worsened over three days. She reported a positive SARS-CoV-2 antigen test from the previous week.

On clinical evaluation, she had a patent airway, was polypneic (respiratory rate of 25 breaths per minute) in ambient air, with an oxygen saturation (SpO_2) of 84% in the seated position. Lung auscultation revealed preserved and symmetrical vesicular breath sounds bilaterally, without adventitious sounds. Other findings included a heart rate of 82 beats per minute, blood pressure of 132/63 mmHg, apyrexia, and a conscious, oriented state without objective neurological deficits. During observation in the emergency department, she remained eupneic at rest in ambient air, with SpO_2 >95%. However, upon standing, she experienced dyspnea and desaturation, requiring increasing oxygen therapy. She was subsequently admitted to the Intensive Care Unit.

Laboratory tests showed hemoglobin of 14.1 g/dL (no anemia), C-reactive protein of 0.40 mg/dL, procalcitonin of 0.04 ng/mL (no signs of infection), B-type natriuretic peptide of 684 pg/mL, and normal thyroid function (thyroid-stimulating hormone 3.042 µUI/mL and free thyroxine T4 1.0 ng/dL). Tests for influenza A, influenza B, and respiratory syncytial virus were negative, as were blood and urine cultures. Electrocardiography revealed regular sinus rhythm with a heart rate of 78 beats per minute and no conduction abnormalities. Computed tomography angiography of the chest excluded pulmonary embolism, pleural effusion, parenchymal consolidations, or ground-glass opacities.

Given persistent hypoxemia, noninvasive ventilation (NIV) was initiated, providing clinical stability in the supine position. However, a postural oxygen saturation difference was noted, with a drop in SpO_2 >5% in the seated position compared to the supine position. Transthoracic echocardiography with bubble test revealed a large right-to-left shunt with a significant volume of bubbles crossing in the first cardiac cycle, confirmed by transesophageal echocardiography. The shunt originated from a large atrial septal aneurysm.

The patient underwent implantation of a 30-mm Atriasept – CARDIA™ prosthesis, which was properly positioned, excluding the aneurysm and resulting in immediate resolution of hypoxia. After 12 months of follow-up, the patient remained free from recurrent dyspnea in the upright position, with no peripheral desaturation in this posture. Transthoracic echocardiography showed no residual shunt.

3. Discussion and Conclusion

The diagnosis of Platypnea-Orthodeoxia Syndrome (POS) is predominantly clinical. Although the degree of hypoxemia may vary, diagnostic criteria include a decrease in arterial oxygen pressure greater than 4 mmHg or in oxygen saturation greater than 5% in the upright position [3, 7]. Transthoracic echocardiography with Doppler is the most useful test for screening right-to-left shunts due to its wide availability, low cost, safety, and sensitivity. However, transesophageal echocardiography is better suited for evaluating atrial and septal anatomy, the inferior vena cava, and the aorta, as well as the dynamic interactions between these structures and their positional variations. The bubble test aids in confirming this condition and distinguishing the etiology of POS: if the bubble passage is observed within the first three cardiac cycles, an intracardiac cause is likely; if only observed after three cycles, an intrapulmonary cause is more probable.

Given that the bubble test is not routinely performed during echocardiograms, this case highlights the importance of considering the diagnostic hypothesis of POS to ensure this specific evaluation is carried out. The gold standard for diagnosing POS is cardiac

catheterization, which demonstrates a discrepancy in oxygen saturation between the pulmonary vein and the aortic artery. However, in daily clinical practice, non-invasive tests are generally sufficient [5, 6, 8].

Thus, the authors emphasize the need for a high index of suspicion in diagnosing this condition, which should be considered whenever patients present with unexplained dyspnea and peripheral oxygen desaturation in the upright position. In such situations, it is recommended to assess peripheral oxygen saturation using pulse oximetry and perform arterial blood gas analysis in both the supine and upright positions, comparing the results. Additionally, echocardiography with a bubble test should be considered.

The treatment of POS depends on its cause. In cases involving intracardiac shunts, as in the case presented, definitive therapy consists of surgical or percutaneous closure of the primary defect. Currently, the preferred approach is percutaneous closure, which offers advantages such as faster post-procedural recovery and lower cost. After treatment, symptoms resolve in 95% of patients, and upright SpO_2 improves. However, in 5% of cases, closure is ineffective due to residual shunts [4, 7, 9, 10]. Therefore, POS represents a rare but potentially treatable cause of respiratory failure. It is essential for clinicians to remain alert to this diagnosis, as treatment can significantly improve the patient's quality of life – as demonstrated in the present case, where hypoxia resolved immediately after surgery in a patient previously dependent on NIV. The authors note that the improvement of symptoms with NIV is an atypical presentation of the disease, which complicated the diagnosis. However, given the strong clinical suspicion raised during ICU admission, further investigation confirmed the diagnosis.

It is worth noting that, despite the patient testing positive for SARS-CoV-2 antigen in the previous week, chest computed tomography did not reveal pulmonary parenchymal abnormalities. In cases of POS associated with COVID-19 described in the literature, pulmonary changes related to the disease were consistently observed. Thus, the authors consider that COVID-19 was not the etiology of POS in this case, although they acknowledge the possibility that the virus acted as a trigger.

Although reported cases of POS seem to have increased in recent years, it is likely that its diagnosis remains significantly underestimated. This hypothesis should be considered by physicians, particularly in the presence of unexplained or paroxysmal hypoxia. The diagnosis is straightforward, and treatment can be curative, resulting in a marked improvement in the patient's autonomy and quality of life [4].

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