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Platypnea-orthodeoxia syndrome in patients presenting enlarged aortic root: case report and literature review

Síndrome de platipneia-ortodeóxia em aorta ectásica: relato de caso e revisão da literatura

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ABSTRACT

We describe herein a case of a patient who, when in orthostatic positions, had severe hypoxemia and ventilatory dysfunction. Although the severity of symptoms required hospitalization in an intensive care setting, the initial tests only identified the presence of enlarged aortic root, which did not explain the condition. The association of these events with an unusual etiology, namely

intracardiac shunt, characterized the diagnosis of platypnea-orthodeoxia syndrome. The literature review shows that, with advancing research methods, there was a progressive increase in the identification of this condition, and this association should be part of the differential diagnosis of dyspnea in patients with enlarged aortic root.

Keywords: Aortic diseases; Respiratory insufficiency; Heart septal defects; Aortic aneurysm; Case reports

INTRODUCTION

Dyspnea and severe hypoxemia exacerbated by orthostatic position are rare events, often related to pulmonary disease or intracardiac shunts.^(1,2) This report describes the research and diagnosis of an unusual etiology: intracardiac shunt secondary to acquired anatomic factors, characterizing platypnea-orthodeoxia syndrome.

CASE REPORT

The patient is female, 86 years old, complaining of dyspnea, productive cough and dorsal thoracic pain, not ventilator-dependent and without radiation, with a progressive condition for more than 7 days. The patient presented with 89% saturation in room air, with no changes upon physical examination. The laboratory tests were normal, as were her electrocardiogram and her chest X-ray. The initial diagnosis was respiratory infection, which was treated with antibiotic therapy. After stabilization, the patient was transferred to the inpatient unit.

At the hospital, the patient suffered hypoxemic respiratory failure, which required a transfer to the intensive care unit (ICU) and the initiation of invasive mechanical ventilation. A chest computerized tomography (CT) and pulmonary CT angiography were performed, which showed no pulmonary embolism and no alteration that justified the condition. The investigation continued with transthoracic echocardiography, which identified a hypertrophic left ventricle, ejection fraction of 68%, changed relaxation pattern, increased left atrial size (4.7mm), an enlarged aortic root with an inner diameter of 45mm, and mild

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aortic and tricuspid valve regurgitation, with an estimated right ventricular/right atrial gradient of 35mmHg. The diagnostic hypothesis was a decompensated diastolic heart failure due to an infection. The patient progressed with rapid improvement and was discharged from the ICU.

The investigation continued, and the patient underwent cardiac catheterization, which showed normal coronary arteries and ruled out pulmonary arterial hypertension.

However, after 48 hours, the patient had a new episode of hypoxic respiratory failure, which was managed in the ICU with noninvasive mechanical ventilation. At that time, the marked worsening of hypoxemia and ventilatory dysfunction that occurred when the patient was sitting or in a more upright position drew attention. This worsening was confirmed through arterial blood gas analyses in both positions, revealing marked orthostatic desaturation (Table 1).

Table 1 - Results of sequential arterial blood gas analyses collected with the patient in different positions

	Supine position	Sitting position
PaO ₂ (mmHg)	116	65
Oxygen saturation (%)	100	96
O ₂ concentration	0.4	0.4

When the possibility of hypoxemia secondary to a right-to-left shunt was suggested, a transesophageal Doppler echocardiography (TEE) was performed with the patient in the supine position with the head tilted 45°. In addition to the finding of enlarged aortic root, this examination revealed the presence of an atrial septal aneurysm and a patent foramen ovale (PFO) with significant and early microbubble flow from the right to the left atrium. During the evaluation, we observed that this flow increased significantly when the head of the patient was elevated (Figure 1).

After the diagnosis of platypnea-orthodeoxia syndrome, the patient underwent percutaneous PFO closure. The patient exhibited good clinical progress and was discharged with no symptoms seven days after the procedure.

DISCUSSION

We report herein the clinical case of a patient with postural accentuation of a right-to-left intracardiac shunt, characteristic of platypnea-orthodeoxia syndrome, characterized by dyspnea and hypoxemia exacerbated or



Figure 1 - Transesophageal echocardiography. Atrial septal aneurysm with high permeability at microbubble contrast.

triggered by orthostasis. This syndrome is caused by a limited number of clinical conditions that produce pulmonary arteriovenous shunt (arteriovenous malformation and hepatopulmonary syndrome) or right-to-left intracardiac shunt (PFO and/or related atrial septal defects).^(1,2)

The prevalence of this syndrome is underestimated because it requires a high degree of suspicion to be identified. However, relatively simple tests (comparison of arterial blood gas analyses performed with the patient in different positions) can confirm the diagnosis, combined with a correct evaluation of the clinical information.^(1,3) Moreover, the possibility of a right-to-left intracardiac shunt should be evaluated by a transesophageal echocardiography with contrast in the supine and upright position. This procedure is a minimally invasive option and provides relatively sensitive diagnosis.^(1,3,4)

PFO is usually asymptomatic because the left atrial pressure tends to keep it closed, through a valve mechanism of the *septum secundum*, and is found in up to 30% of the adult population.⁽⁵⁾ However, in situations of increased pressure in the right heart chambers or redirected venous flow toward the face of the PFO, aimed at the right atrium, right-to-left shunt may occur. When a shunt occurs due to changes in the pressure gradient (higher pressure in the right atrium than in the left), neither dyspnea nor hypoxemia is influenced by the position of the patient.^(4,5)

In platypnea-orthodeoxia syndrome, right-to-left shunt is not related to increased pressure in the right heart chambers. For shunt to occur, other factors must be present to change the intracardiac anatomical relationships and to orient the venous flow toward the atrial septum, causing

the venous blood to cross the PFO toward the left atrium. Thus, atrial shunt is caused or potentiated by orthostatism, manifesting in platypnea-orthodeoxia complaints.^(2,4-7)

There are two groups of proposed anatomical factors that, by coexisting with PFO, may determine intracardiac shunt with normal pressure, namely congenital (persistence of fetal circulation) and acquired factors. Examples of congenital factors include the Chiari network and the redundant Eustachian valve (present in the anterior inferior face of the inferior vena cava). The following acquired factors have been described: pneumonectomy, atelectasis, diaphragm paralysis, kyphoscoliosis, and enlarged aortic root/aneurysm.^(2,4-8) In theory, the mediastinal remodeling that occurs with acquired factors would change the conformation (tending to stretch it) and the position of the atrial septum, favoring the passage of blood from the right to the left side in the presence of PFO. Thus, orthostatism would contribute to directing the blood flow from the inferior vena cava to the atrial septum, thereby extending the shunt.^(2,4-9)

An association between enlarged aortic root and the development of right-to-left shunt has been described in various populations.^(2,5,7-9) In 2005, Eicher et al. retrospectively evaluated a series of 19 patients with platypnea-orthodeoxia syndrome due to intracardiac shunt and found aortic root dilation in 63% of them.⁽⁸⁾ The researchers also revealed that in 42% of cases, there was no other thoraco-pulmonary change that could justify the syndrome; thus, enlarged aortic root was the only likely etiological agent.⁽⁸⁾ In 2007, Bertaux et al. described changes in the conformation, size and mobility of the atrial septum determined by the presence of aortic root aneurysm in a prospective study.⁽⁹⁾ Bertaux et al.

found that the higher the dilation of the aortic root, the smaller and more mobile the atrial septum becomes; they also found that in the presence of PFO, this change would potentiate (proportionally to the ectasia size) the right-to-left shunt.⁽⁹⁾

Platypnea-orthodeoxia syndrome greatly affects the quality of life of patients. Thus, repairing the atrial septal defect is essential for the relief of symptoms. In the literature, both percutaneous and surgical treatments are indicated to be effective. However, percutaneous PFO closure is associated with high success rates when implanting the prosthesis and with a low risk of complications.⁽¹⁰⁾ This technique may be more highly recommended in some situations, such as in the case of our patient, because it avoids the morbidity and mortality and higher costs associated with open heart surgery.

CONCLUSION

Platypnea-orthodeoxia syndrome secondary to a right-to-left shunt in the absence of increased pressure in the heart chambers is a presumably underdiagnosed event. We emphasize that one of the proposed pathophysiological mechanisms, namely enlarged aortic root, has a high prevalence and tends to increase progressively. Thus, platypnea-orthodeoxia syndrome should be considered in the differential diagnosis of causes for hypoxemia and ventilatory dysfunction in patients with enlarged aortic root.

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RESUMO

Descrevemos aqui o caso de um paciente que, ao assumir posições de ortostatismo, apresentava hipoxemia e disfunção ventilatória grave. Embora a gravidade dos sintomas tenha determinado a necessidade de internação em ambiente de terapia intensiva, os exames iniciais identificaram apenas a presença de ectasia da aorta, sem, no entanto, justificar o quadro. A associação dessas manifestações a uma etiologia incomum, o shunt

intracardíaco, caracterizou o diagnóstico da síndrome de platipnea-ortodeóxia. A revisão da literatura demonstra que, com o avanço dos métodos de investigação, houve progressivo aumento na identificação desse quadro, devendo essa associação fazer parte do diagnóstico diferencial de dispnéia em pacientes com aorta ectásica.

Descritores: Doenças da aorta; Insuficiência respiratória; Defeitos do septo cardíaco; Aneurisma aórtico; Relatos de casos

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