Rare disease

Platypnoea-orthodeoxia syndrome

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Summary

A 78-year-old man with a history of gastric ulcer and pulmonary embolism was admitted for elective revision of a right total hip replacement. He was mildly hypoxic preoperatively (saturation 89% on air). He became profoundly breathless postoperatively (saturation 75%). He was treated for presumed pulmonary oedema but failed to improve. A CT pulmonary angiogram and transthoracic echo showed no clear cause for his symptoms. Because the patient's symptoms were postural, exacerbated in the upright position and relieved by lying supine, the authors suspected a diagnosis of platypnoea-orthodeoxia syndrome associated with a patent foramen ovale (PFO). Transoesophageal echo and microbubble study confirmed he had a PFO. The patient's PFO was percutaneously closed and his symptoms and positional hypoxia completely resolved.

BACKGROUND

Platypnoea-orthodeoxia syndrome (POS) is a rare and complex phenomenon which is not fully understood. We describe an unusual case of POS that first presented postoperatively and was associated with a patent foramen ovale (PFO), aortic root dilation and interatrial septal aneurysm. The diagnosis of POS is challenging and requires a high degree of clinical suspicion. POS is potentially curable and it is, therefore, important that clinical awareness of the syndrome is raised among practicing clinicians.

CASE PRESENTATION

A 78-year-old man with a history of gastric ulcer and pulmonary embolism (PE), was admitted for elective revision of a right total hip replacement. A routine echocardiogram in 2009 was normal.

On admission, he was mildly hypoxic (arterial oxygen saturation 89% on air). He was asymptomatic and a preoperative chest radiograph was normal, so surgery went ahead as planned. Postoperatively, the patient complained of chest tightness and shortness of breath, and became increasingly hypoxic (arterial oxygen saturation 75%). ECG was unremarkable and troponin T was only mildly elevated at 22 ng/l. On examination, he had widespread wheeze and was treated for presumed pulmonary oedema with morphine, nitrates and furosemide. He failed to improve and required continuous positive airway pressure (CPAP) and inotropic support and was transferred to the intensive care unit (ICU).

The patient became transiently febrile and was treated for possible hospital-acquired pneumonia with intravenous antibiotics. A diagnosis of PE was also considered because of his significant risk factors (prior history of PE and recent major surgery). However, a CT pulmonary angiogram (CTPA) was normal and revealed no cause for his symptoms.

After 2 days, the patient improved and was transferred to a surgical ward, but continued to have episodes of profound shortness of breath which appeared to worsen when supine. Ten days into rehabilitation while attempting to stand with the physiotherapist, the patient collapsed and became briefly unresponsive with an arterial oxygen saturation of 81%, and was re-admitted to the ICU.

A further CTPA was normal. The cardiologists recommended a transthoracic echo (TTE) and 24-h ECG, which showed no significant abnormalities and the diagnosis remained unclear.

Despite exclusion of the main causes of hypoxia, the patient's symptoms persisted and he was transferred to the cardiology department for further investigation. We noticed that the patient's symptoms were directly related to posture. His arterial oxygen saturation when lying flat was normal but fell to 75% on sitting, confirmed by arterial blood gas. We suspected a diagnosis of POS associated with a PFO.

Cardiac catheterisation demonstrated minor plaque disease, mild-to-moderate aortic regurgitation, a mildly dilated aortic root (41 mm) and a probable communication between the right and left atria, but could not confirm a PFO. Right-sided heart pressures were normal with no evidence of pulmonary hypertension.

Transoesophageal echocardiogram (TOE) revealed an interatrial septal aneurysm with a large PFO. The patient tolerated the procedure poorly and was unable to perform a change in position to demonstrate the PFO clearly. The test did, however, induce a Valsalva effect which enabled the PFO to be more easily seen. The PFO was confirmed on microbubble testing with transthoracic echocardiography (figure 1).

The PFO was successfully closed at a tertiary referral centre. His positional dyspnoea and hypoxia completely resolved, confirming the diagnosis of POS.

Figure 1 Transthoracic echo with microbubble study. Microbubbles flow from the right atrium (RA) through the patent foramen ovale in the interatrial septum (IAS) into the left atrium (LA) and then left ventricle (LV). Normally, microbubbles would only be seen in the right-sided heart chambers.

INVESTIGATIONS

- Preoperative arterial blood gas on air: PaO₂ 9.24 kPa, PaCO₂ 4.73 kPa, pH 7.412 and base excess −2.7.
- Postoperative arterial oxygen saturation recumbent: 96%.
- Postoperative arterial oxygen saturation when upright: 75%.
- Chest radiograph: lung fields clear.
- ECG: first-degree heart block. Otherwise normal.
- CTPA (1) and (2): no evidence of pulmonary emboli and no other significant abnormalities were seen.
- TTE: good left and right ventricular function with a mobile intra-atrial septum. Mild central aortic regurgitation.
- Twenty-four hour ECG: sinus rhythm with first degree heart block throughout with frequent multifocal premature ventricular complexes.
- Cardiac catheterisation: minor plaque disease. Grade 2–3 aortic regurgitation. Dilated aortic root (41 mm). Probable communication between right and left atria, but a PFO could not be confirmed. No evidence of pulmonary hypertension.
- TOE: interatrial septal aneurysm. Large PFO with right to left shunt seen with microbubble testing.

DISCUSSION

POS was first described in 1949 by Burchell *et al*¹ and is defined by the development of dyspnoea and hypoxia while in the upright position which improves once recumbent. There are fewer than 100 cases reported in the literature.² It is caused by various cardiac, pulmonary and hepatic conditions and is usually associated with an intracardiac or intrapulmonary right to left shunt.

Two conditions must co-exist for the syndrome to manifest: an anatomical and a functional component.³ In

cardiac POS, the anatomical component usually comprises an interatrial communication causing a right to left shunt, most commonly a PFO. Rarer examples include atrial septal defects and septal aneurysms. The functional component allows a redirection of blood flow via the shunt when the patient changes position from recumbent to upright, causing positional hypoxia and dyspnoea. Reported functional elements include constrictive pericarditis or pericardial effusion, pneumonectomy or lobectomy, aortic aneurysm or elongation and compression of the chambers of the right heart by liver lesions.⁴ Pulmonary hypertension is generally absent.

A PFO can be identified in approximately 27% of the population,⁴ however, POS is rare. The key to understanding the development of POS, therefore, lies in identifying the functional component. Interestingly, in our case, this may have been multifactorial. First, dilation of the aortic root may have deformed the atrial septum enough on standing to widen the PFO and exacerbate right to left shunting, resulting in paradoxical hypoxia on sitting. This dilation was likely to have been degenerative in nature, which would explain the late onset of the syndrome in this patient. Second, increased right-sided heart pressure during intubation and CPAP may have widened the PFO and made the shunting more apparent postoperatively. This appears to be the first reported case in which ventilation has exacerbated POS.

This case illustrates that the diagnosis of POS is challenging, and requires a high degree of clinical suspicion. Key points in diagnosis include demonstrating hypoxia in the upright position, which improves on lying flat and detecting a right to left shunt. The most sensitive non-invasive test to confirm the latter is TOE with bubble study.² Manoeuvres that increase the right to left shunt, such as the Valsalva manoeuvre, can be used to aid the diagnosis.

Although difficult, identification of this syndrome is crucial because prompt diagnosis and treatment can be curative.

Learning points

- ▶ POS is a rare cause of hypoxia and dyspnoea.
- ▶ The key feature of POS is that symptoms are relived by lying supine.
- ▶ POS comprises of both a functional and anatomical component (often a PFO).
- ▶ The diagnosis is challenging.
- ▶ POS is potentially curable.

Competing interests None.

Patient consent Obtained.

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