Platypnoea–orthodeoxia syndrome due to deformation of the patent foramen ovale caused by a dilated ascending aorta: a case report

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Received 9 October 2019; first decision 1 November 2019; accepted 28 January 2020

Background
Platypnoea–orthodeoxia syndrome (POS) is characterized by dyspnoea and arterial desaturation in the sitting position. Although its pathophysiology is complex and still needed to be investigated, the disease is one of the clinical situations which should be immediately and adequately managed by health care workers from the initial presentation.

Case summary
A 66-year-old woman with a history of systemic lupus erythematosus, deep vein thrombosis, and lumbar compression fracture was admitted for evaluation of the sudden onset of dyspnoea, while in the sitting position that was relieved on placing her in the supine position. Her transoesophageal echocardiogram did reveal a deformity in the patent foramen ovale (PFO) structure with a wide gap due to aortic compression, which was markedly different from that observed in the supine position, along with massive right-to-left shunting caused by redirected venous return due to a persistent Eustachian valve. With the computed tomography and angiograms, POS was diagnosed. Then, the patient received aortic replacement and patch closure of PFO, and her symptoms were completely resolved.

Discussion
Platypnoea–orthodeoxia syndrome is a condition with quite unique features and needs multiple clinical measures for the diagnosis and medical management. For all health care workers, it is essential to have a high suspicion in order to detect POS in patients with unexplained dyspnoea. Echocardiography plays a major role in establishing the diagnosis and offering the choice of therapeutic options.

Keywords
Case report • Echocardiography • Patent foramen ovale • Orthodeoxia

Learning points
• To show typical pathophysiology of platypnoea–orthodeoxia syndrome (POS) in the elderly.
• To highlight the role of echocardiography to detect and to manage POS.
Introduction

Platypnoea–orthodeoxia syndrome (POS), which is characterized by dyspnoea and arterial desaturation in the sitting position, was believed to be a rare clinical entity. The proposed pathophysiology is complex and involves the following two components: an anatomic component, such as an interatrial communication, and a secondary or functional component that results in a deformation in the atrial septum with redirection of shunt flow in the upright posture.

Timeline

| Date     | Events |
|----------|--------|
| 2001     | Diagnosis of systemic lupus erythematosus |
| June 2018| Admission to the prior medical institution due to bone fracture of the pelvis |
| 2 July 2018| Progression of orthostatic dyspnoea |
| 2 July 2018| Transferred to our institution |
| 3 July 2018| Electrocardiogram, chest X-ray, and transthoracic echocardiography |
| 4 July 2018| Not remarkable |
| 4 July 2018| Computed tomography |
| 4 July 2018| Ascending aorta dilation |
| 6 July 2018| Comprehensive right heart catheterization including right atrial angiogram |
| 6 July 2018| Existence of the right to left shunt at the level of atriaums |
| 6 July 2018| Transoesophageal echocardiography |
| 12 July 2018| De novo demonstration of the augmented right to left shunt through patent foramen ovale (PFO) by sitting position |
| 12 July 2018| Surgical repair of the PFO and resection of ascending aortic aneurysm |
| 17 July 2018| No relapse of the symptoms |
| 30 July 2018| Transoesophageal echocardiography |
| 30 July 2018| No residual intracardiac shunt |
| 30 July 2018| Discharge |

Case presentation

A 66-year-old woman with a history of systemic lupus erythematosus, deep vein thrombosis, and lumbar compression fracture was admitted for the evaluation of sudden onset dyspnoea, while in the sitting position that was relieved when lying supine. She had become frail as a result, having spent most of the day lying in bed due to the above-mentioned symptoms. As long as she was recumbent, her condition was stable, and her physical examination did not reveal any cardiopulmonary signs. Her general laboratory results were within the normal limits, except for a relatively high brain natriuretic peptide value which was 35.4 pg/mL (0–18.4 pg/mL). The electrocardiogram was normal, although her chest X-ray revealed mild dilatation of the upper mediastinum. However, in the sitting position, her blood gas analysis revealed marked hypoxaemia with low partial pressure of arterial oxygen (PaO2) and oxygen saturation (54.1 mmHg and 75%, respectively) in room air, which quickly recovered to the normoxic status with normal PaO2 and oxygen saturation (102.6 mmHg and 100%, respectively) in the supine position. While transoesophageal echocardiogram showed preserved left ventricular ejection fraction (67%) and failed to detect any specific findings, her transoesophageal echocardiogram (TOE) revealed a patent foramen ovale (PFO) with a positive microbubble test, but without obvious right-to-left shunting in the supine position (Figure 1A, Supplementary material online, Videos S1 and S2). In the sitting position, TOE further revealed a deformity in the structure of the PFO with a wide gap due to aortic compression that was markedly different from that observed in the supine position. Massive right-to-left shunt flow caused by redirected venous return could be observed due to the persistent Eustachian valve (Figure 1B and Supplementary material online, Video S3). Therefore, POS was diagnosed. Latterly, the maximum diameter of the ascending aorta was measured to be 57 mm on contrast-enhanced computed tomography (Figure 1C). The elongated ascending aorta expanded to pressurize the right atrium. Finally, the presence of an intra-atrial shunt was confirmed on the right atrial angiogram (Supplementary material online, Video S4). After the heart team conference, she received surgical prosthetic vascular graft replacement of the ascending aorta and patch closure of PFO in the supine position under cardiopulmonary bypass. Immediately after the surgery, her blood gas analysis normalized even in the sitting position, and her symptoms disappeared completely. A post-operative TOE revealed no residual intracardiac shunt in the sitting position (Figure 1D and Supplementary material online, Video S5). She was discharged without any events, and no recurrence occurred up to the present.

Discussion

In this patient, the anatomical component was the PFO, and the secondary component included both cardiac factors, such as the existence of a persistent Eustachian valve, and vascular factors such as a dilated aortic root. Agrawal et al. conducted a full review of the literature from 1949 to 2016 regarding POS and reported PFO as the most common anatomical component. Since PFO was detected in 27% of the autopsy cases of the general population, it can be considered a relatively common condition. Recently, several case reports have documented the contribution of aortic atherosclerosis, which led to elongation of the ascending aorta or aortic root dilation as the secondary component of POS in the elderly population. As the rate of atherosclerosis that induces aortic elongation or aortic root dilatation increases in the population annually, the combination of congenital and acquired pathologies (PFO and aortic compression) may now be considered one of the typical features of POS.

For senior patients who are complicated due to multiple health problems and who are otherwise prone to have a greater need for bed rest, establishing the diagnosis of POS can be challenging. These patients sometimes cannot explain or easily express their pattern of symptoms, and extensive use of laboratory and diagnostic imaging modalities is required to rule out several similar health conditions.
Measuring arterial blood gases in different positions and obtaining the expected results from positional change shall be the initial step for the successful diagnosis of POS. Subsequently, contrast-enhanced transthoracic echocardiogram or TOE will confirm the existence of right-to-left intracardiac shunt and uncover the mechanism. Once the diagnosis is established, therapeutic options are usually deployed because surgical or percutaneous treatments have been already established. And, depending upon the patient’s overall health condition, the symptoms are then usually relieved.

**Conclusion**

POS is a condition with quite unique features and needs multiple clinical measures for the diagnosis and medical management. Among them, echocardiography plays a major role in establishing the diagnosis and offering the choice of therapeutic options. As the number of POS cases is growing in the elderly population, POS may be one of the emerging health risks of ageing in Western societies such as Europe, the UK, the USA, and Japan. For physicians, it is essential to have a high suspicion in order to detect POS in patients with unexplained dyspnoea, especially in the elderly population.

**Lead author biography**

Dr Misaki Hasegawa was born in Yokkaichi-shi, Japan in 1988. She received the MD degree from Tokai University School of Medicine (Isehara-shi, Japan) in 2015. Since 2018, she has been enrolled in the PhD programme of cardiovascular medicine (Tokai University Graduate School of Medicine, Isehara-shi, Japan). Her research interest is mainly focused on cardiac imaging in coronary artery diseases and heart failure.

**Supplementary material**

Supplementary material is available at *European Heart Journal - Case Reports* online.

**Slide sets:** A fully edited slide set detailing this case and suitable for local presentation is available online as Supplementary data.
Consent: The author/s confirm that written consent for submission and publication of this case report including image(s) and associated text has been obtained from the patient in line with COPE guidance.

Conflict of interest: The authors declare no conflict of interest.

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