Reminder of important clinical lesson

If at first you do not succeed, think again!

Subramanya G N Upadhyaya,¹ Andrew J Ritchie,² Toomas Sarev,¹ Alisdair Ryding¹¹Cardiology Department, James Paget Hospital, Great Yarmouth, UK;²Cardiothoracic Surgery Department, Basildon and Thurrock University Hospital, Basildon, UK**Correspondence to** Dr Alisdair Ryding, Alisdair.Ryding@jpaget.nhs.uk**Summary**

The authors describe a case of platypnoea orthodeoxia syndrome in an 83-year-old man with a fenestrated atrial septal defect and severe coronary artery disease. The patient had been admitted to hospital six times in the previous year with acute breathlessness, attributed to paroxysmal atrial fibrillation. The patient's symptoms resolved completely following surgical repair of the defect and coronary artery bypass grafting.

BACKGROUND

Platypnoea orthodeoxia syndrome is an extremely rare cause of acute breathlessness, requiring a high index of suspicion and careful investigation to reach a diagnosis. This case is unusual for prolonged delay in diagnosis, the advanced age at presentation and the presence of a fenestrated atrial septal defect (ASD).

CASE PRESENTATION

An 83-year-old gentleman presented with a history of worsening breathlessness and dizziness for 4 days. He was known to have paroxysmal atrial fibrillation (PAF) and had been admitted to same hospital with similar symptoms 6 times in the past 12 months. On each previous occasion, he was discharged home with the diagnosis of PAF, and optimisation of antiarrhythmic medication. He had undergone ambulatory ECG monitoring without any evidence of significant arrhythmia detected. Other medical history includes acute myocardial infarction in 1990, coronary balloon angioplasty in 1995, hypertension, chronic kidney disease stage 3b and mild short term memory impairment.

His regular medications included bisoprolol 2.5 mg, warfarin, simvastatin 40 mg, omeprazole 20 mg, citalopram 10 mg, tamsulosin 400 mcg, nicorandil 10 mg twice daily and isosorbide mononitrate 20 mg once daily. Clinical examination demonstrated a regular pulse at 57 bpm, blood pressure 121/75 mm Hg and oxygen saturations of 96% on room air. Mini mental test was 8 out of 10. There were no abnormal physical findings. Routine bloods including full blood count, D-dimer and troponin were normal. Chest x-ray was unremarkable. ECG showed sinus bradycardia.

The next day the patient developed acute onset respiratory distress while sitting in a chair. Oxygen saturations dropped to 70% on air and arterial blood gas sampling revealed severe type I respiratory failure resistant to high flow oxygen. During this he remained in sinus rhythm. Eventually the patient recovered spontaneously in the recumbent position. For the remainder of the admission the

patient was unable to sit up without rapidly desaturating and becoming profoundly unwell.

INVESTIGATIONS

ECG: sinus rhythm 57 bpm, normal axis, normal QRS complexes.

Arterial blood gases (ABG) sitting upright: pO₂ 4.5 kPa, pCO₂ 3.65 kPa on 2 litres O₂/min. ABG supine: pO₂ 11.2 kPa on room air.

CT pulmonary angiography: normal.

Transthoracic echocardiography (left lateral decubitus position): normal, though suboptimal views.

Transoesophageal echocardiography (left lateral decubitus position): this revealed an atrial septal aneurysm with colour flow Doppler confirming a fenestrated secundum ASD with left to right shunt (figure 1).

Cardiac catheterisation (supine): right atrial pressure: a wave 9 mm Hg, v wave 0 mm Hg, mean 1 mm Hg.

Right ventricular pressure: systolic 20 mm Hg, diastolic 0 mm Hg, end diastolic 0 mm Hg.

Pulmonary artery pressure: systolic 19 mm Hg, mean 14 mm Hg.

Pulmonary capillary wedge pressure: a wave 2 mm Hg, v wave 0 mm Hg, mean 0 mm Hg.

Left ventricular pressure: systolic 117 mm Hg, diastolic 0 mm Hg, end diastolic 3 mm Hg.

A small right to left shunt was evident in the supine position (Fick method: Qp:Qs=0.8).

Coronary angiography: significant distal left main stem stenosis and proximal three-vessel coronary artery disease (figure 2).

DIFFERENTIAL DIAGNOSIS

Platypnoea orthodeoxia syndrome secondary to a fenestrated secundum ASD and severe coronary artery disease.

TREATMENT

Surgical patch closure of the fenestrated ASD and coronary artery bypass grafting was undertaken. Intraoperative photographs clearly demonstrate the fenestrated nature of the ASD (figure 3).

Figure 1 Transoesophageal echocardiography. A) Interatrial septal aneurysm. B) Fenestrated atrial septal defect with left to right shunting.

OUTCOME AND FOLLOW-UP

Surgery resulted in complete resolution of the patient's symptom of orthodeoxia when followed up at 3 months, although he had further admissions with PAF.

DISCUSSION

This is a case of platypnoea orthodeoxia syndrome (POS) due to late manifestation of a fenestrated secundum ASD and atrial septal aneurysm. It is a rare condition characterised by profound desaturation in the upright position (orthodeoxia), with resolution in the supine position (platypnoea).¹ It was first described in 1949 by Burchell *et al*,² and since then around a further 100 cases have been reported in the literature. The case described in this report is unusual for the very advanced age at presentation, the presence of a fenestrated ASD, and the association with advanced coronary artery disease.

The commonest cause is a patent foramen ovale or secundum ASD with right to left shunting in the upright position.¹ Fenestrated ASD seems to be an unusual cause, having been reported only twice before.^{3 4} Extra-cardiac shunts have also been described (eg, pulmonary arteriovenous malformations).¹

Diagnosis of this condition is often delayed due to the intermittent and unusual pattern of symptoms and signs, requiring a high degree of suspicion to prompt and interpret appropriate investigations. In this case transthoracic echocardiography was non-diagnostic and transoesophageal echocardiography was required to demonstrate a

Figure 2 Coronary angiography. A) Left coronary system. Right anterior oblique caudal view. B) Right coronary artery. Left anterior oblique view.

fenestrated ASD that could be the substrate for POS. In the left lateral decubitus position left to right shunting was evident on TOE: demonstration of shunt reversal on sitting up was not practicable during this test. Unexpectedly cardiac catheterisation suggested a small right to left shunt in the supine position, which was presumed to worsen in the upright position. The reason for this discrepancy in shunt direction between different tests is difficult to explain, but probably reflects the complex interplay of factors that determine the direction of blood flow in these cases, as discussed below.

The development of a right to left interatrial shunt can be easily explained when right heart pressure is elevated (eg, pulmonary embolism, right ventricular infarction, pneumonectomy, chronic lung disease).¹ However, when right-sided pressures are normal/low, such as this case, the direction of the shunt is probably determined by a complex mixture of factors. First, small interatrial pressure gradients may transiently exist due to the differential effects of respiration on left and right atrial filling. Such gradients are difficult to measure during cardiac catheterisation in the supine position, and impossible to measure in an upright position. Second, anatomical factors have been identified that direct blood flow towards the septal defect (eg, prominent

Figure 3 Intraoperative photographs. A) Inter atrial septum with fenestrated atrial septal defect held with forceps. B) Atrial septal defect patch closure.

Eustachian valve/Chiari network),⁵ or stretch the interatrial septum open (eg, abnormal aortic root position, aortic aneurysm, kyphoscoliosis, lipomatous hypertrophy of the interatrial septum, atrial septal aneurysm).^{1–6} These anatomical factors therefore promote right to left shunting, and provide some explanation for the positional dependency of POS.

The mean age at presentation in currently published cases is 66 years, making this case one of the oldest reported to date. In addition to the underlying congenital ASD, this patient had PAF and severe ischaemic heart

disease. Although episodes of desaturation were unrelated to episodes of atrial fibrillation or angina, it is possible that these pathologies affected atrial structure and function, contributing to the development of POS.

Our case highlights the high index of suspicion required to diagnose POS. In patients with a compatible history and objective evidence of reversible desaturation in an upright position rigorous efforts should be made to identify a shunt. If transthoracic echocardiography is non-diagnostic, transoesophageal echocardiography is mandatory. Furthermore, without an awareness of this condition an apparently small and innocuous interatrial shunt can be easily overlooked.

Learning points

- ▶ Platypnoea-orthodeoxia syndrome is a rare but well-recognised condition and should be considered in patients presenting with postural dyspnoea/desaturation.
- ▶ The cardinal feature is cyanosis in the upright position, with recovery lying flat.
- ▶ A high index of suspicion is essential to obtain the relevant history, and appropriate investigations. Transthoracic echocardiography may not be diagnostic.
- ▶ POS is a treatable either by percutaneous or surgical closure depending on the exact anatomical defect.

Competing interests None.

Patient consent Obtained.

REFERENCES

1. **Cheng TO.** Mechanisms of platypnea-orthodeoxia: what causes water to flow uphill? *Circulation* 2002;**105**:e47.
2. **Burchell HB, Helmholtz HF Jr, Wood EH.** Reflex orthostatic dyspnea associated with pulmonary hypertension. *Am J Physiol* 1949;**159**:563–4.
3. **van Gaal WJ, Joseph M, Jones E, et al.** Platypnea-orthodeoxia associated with a fenestrated atrial septal aneurysm: case report. *Cardiovasc Ultrasound* 2005;**3**:28.
4. **Giombolini C, Notaristefano S, Santucci S, et al.** Platypnea-orthodeoxia induced by fenestrated atrial septal aneurysm. *Ital Heart J* 2005;**6**:164–7.
5. **Shakur R, Ryding A, Timperley J, et al.** Late emergence of platypnea orthodeoxia: chiari network and atrial septal hypertrophy demonstrated with transoesophageal echocardiography. *Eur J Echocardiogr* 2008;**9**:694–6.
6. **Eicher JC, Bonniaud P, Baudouin N, et al.** Hypoxaemia associated with an enlarged aortic root: a new syndrome? *Heart* 2005;**91**:1030–5.

This pdf has been created automatically from the final edited text and images.

Copyright 2012 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Please cite this article as follows (you will need to access the article online to obtain the date of publication).

Upadhyaya SGN, Ritchie AJ, Sarev T, Ryding A. If at first you do not succeed, think again!.
BMJ Case Reports 2012;10.1136/bcr.12.2011.5442, Published XXX

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow

Keep up to date with all published cases by signing up for an alert (all we need is your email address) <http://casereports.bmj.com/cgi/alerts/etoc>