Case report - Congenital

Cyanosis in atrial septal defect without pulmonary hypertension: a case of Platypnea-orthodeoxya syndrome

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Abstract

Cyanosis in atrial septal defect typically occurs when pulmonary hypertension develops. Platypnea-orthodeoxya is an uncommon syndrome, still under debate, characterized by breathlessness and arterial oxygen desaturation exacerbated in the upright position. An interatrial communication is a common finding in this syndrome, but the absence of a right to left pressure gradient complicates the physiopathological picture. To explain the right to left shunt, it is generally advocated a concomitant condition that alters the sterical relationship between inferior vena cava orifice and the atrial septal defect. A case of a 58-year-old male with platypnea-orthodeoxya syndrome related to a fenestrated redundant interatrial septum without any additional pathologic condition is reported. Possibly, this isolated anatomical abnormality could lead to a right to left shunt in the absence of other coexisting predisposing factors. It is reasonable to hypothesize the septum secundum bulging like a ‘spinnaker’ into the right atrium, so that it deviates the inferior vena cava venous blood towards the left atrium. Echocardiographic evaluation is mandatory to achieve a correct diagnosis and to decide the therapeutic strategy. © 2005 Published by European Association for Cardio-Thoracic Surgery. All rights reserved.

Keywords: Atrial septal defect; Intracardiac shunt; Cyanosis

1. Introduction

Hypoxaemia in atrial septal defect (ASD) is generally related to a right to left shunt due to pulmonary vascular disease. Platypnea-orthodeoxya syndrome is a rare condition characterized by breathlessness and arterial oxygen desaturation worsened in the upright position. This clinical condition was first reported by Burchell [1] and is associated with atrial septal defect and right to left shunt in spite of normal right heart pressures. It has been found to be associated with elongated ascending aorta, pneumonectomy, liver cirrhosis, and recurrent pulmonary embolism [2]. We report a case of Platypnea-orthodeoxya syndrome in which a redundant and fenestrated atrial septum was found and it could be involved in the pathogenesis of this syndrome. No additional pathological findings were observed.

2. Case report

A 58-year-old male was admitted to our institution for chronic hypoxaemia with cyanosis, secondary poliglobulia (hematocrit 50%, hemoglobin 20.5 mg/dl, red cell count 6.44X10⁶·u/l), clubbing, peripheral arterial blood saturation 80% in clinostatism decreasing to 67% in orthostatism, and a recent transitory ischaemic attack.

The chest X-ray was normal and the ECG showed first degree atrio-ventricular block and left bundle branch block. Transesophageal echocardiography excluded anomalous venous returns and showed a redundant atrial septum without evident right to left shunt. Injection of saline solution into the inferior vena cava at color-doppler evaluation revealed light right to left echocontrast migration. Haemodinamic evaluation showed right to left shunt (QP/QS=0.89) without pulmonary hypertension (pulmonary artery pressure 28/13 mmHg, mean 18 mmHg), right atrial pressure 9 mmHg, left atrial pressure 11 mmHg and arteriolar pulmonary resistance 1.49 Wood Units. Oxygen saturation was 75% in inferior vena cava, 72% in the right atrium, 97% in the pulmonary veins and 87.6% in the aorta. Intraoperative transesophageal echocardiography confirmed the presence of a redundant septum (Fig. 1a). The surgical procedure was performed on standard cardiopulmonary bypass under cold blood cardioplegic arrest. After opening the right atrium a clearly redundant thin, hyaline septum secundum was found. It was characterized by several small holes in the upper portion (Chiari network, Fig. 1b), while the lower area presented a large defect located just above the orifice of the inferior vena cava (Fig. 2). The Eustachian valve was normal. After removing the excess of interatrial septum tissue the defect was closed by a dacron patch.
After the surgical procedure cyanosis disappeared and arterial oxygen saturation was 100% independently on posture. The postoperative course was uneventful and the patient was discharged home on day seven.

3. Discussion

Right to left shunt in fenestrated atrial septum aneurysm without pulmonary hypertension is a rare condition with relevant complications such as chronic cyanosis and paradox systemic embolism. Platypnea-orthodeoxia is even a rare syndrome whose underlying physiopathologic bases are still under investigation. Approximately 50 years after its first description, in 1949, this syndrome was identified on only about forty occasions [3]. Currently, there is a renewed interest about this syndrome concerning the theoretical explanations of its pathogenesis. There seems to be agreement concerning this syndrome that it is a result of a combination of different pathological conditions [4,5]. Cheng and colleagues [4] categorize these conditions into two groups: anatomical and functional. The first is the presence of an atrial septal defect. The functional is related to the effect of some components that produce a deformity of the interatrial septum such as aortic elongation/aneurysm, pneumonectomy/lobectomy or loculated pericardial effusion resulting in a redirection of shunt flow in the upright posture. None of these functional conditions were present in our case.

Godart and colleagues [6] described two theories to explain right to left shunt with normal pulmonary pressure. The first was related to the presence of a right to left pressure gradient correlated with some clinical conditions like right atrial myxoma, right ventricular infarction or mechanical ventilations. The second was related to the presence of a redundant atrial septum displaced towards the horizontal plane with atrial septal defect and the over-developed Eustachian valve leading to preferential drainage of the inferior vena cava blood flow to the left atrium. Unfortunately, the underlying anatomical pattern could not be demonstrated in this report because the atrial defects were closed percutaneously.

Thomas and colleagues [7] described a case of platypnea-orthodeoxia where the right to left shunt was realized by the presence of a redundant septum secundum associated with an over-developed Eustachian valve.

In the case reported here, the only anatomical abnormal pattern we found was an aneurismal and fenestrated atrial septum with a large defect lying just in front and above the orifice of the inferior vena cava; the Eustachian valve was judged normal. So this single abnormality was enough to cause the deranging of the inferior vena cava desaturated venous blood flow to the left heart and thus to the systemic circulation.

Upright posture, probably by pressure gradient, could increase the stretching of the interatrial septum that could assume a ‘spinnaker’ aspect into the right atrium thus increasing the blood flow to the left direction.

In conclusion, in the presence of cyanosis in atrial septal defect without pulmonary hypertension, a platypnea-orthodeoxia syndrome has to be investigated. If confirmed, it is mandatory to exclude coexisting clinical conditions but should be considered that a single anatomical lesion can be the origin of this complex clinical pattern. A correct diagnosis and surgical treatment can be achieved by matching the morphological and functional data obtained from echocardiography and cardiac catheterization.
References


Appendix A. ICVTS online discussion

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eComment: I have a few ideas to be mentioned in such a case. Platypnea is breathlessness in the upright position and orthodeoxia is arterial desaturation in the upright position. Patients should have confirmation of orthostatic desaturation by erect and supine pulse oximetry. Arterial blood gases performed on room air showed a significant hypoxaemia (PaO2 68 mmHg) with an alveolar to arterial gradient of greater than 40 mmHg. Pulmonary ventilation perfusion scan, non-invasive peripheral vascular studies of the lower limb venous system, and pulmonary function tests were normal. Platypnea-orthodeoxia syndrome [1]. Right-to-left shunting through a patent foreman ovale (PFO) is mostly caused by increased right arterial pressure (massive pulmonary embolism or primary pulmonary hypertension). Another major cause is an abnormal anatomic relationship with a change in the blood flow from the inferior caval vein directed to the PFO. This condition may be seen after pneumonectomy [2]. Right-to-left shunting across a PFO in the absence of lung and heart disease has been reported. Recently, Faller et al [3] described a right-to-left shunt through a PFO in association with both an atrial septum aneurysm and a thoracic aorta aneurysm. Sorrentino et al. [4] described a case of platypnea and orthodeoxia caused by right-to-left shunting across a PFO in a patient with normal pulmonary artery pressure and normal heart function, versus the upright position. Placement of a CardioSEALTM with a percutaneous catheter technique is presently the treatment of choice for this disease [5]. PFO is the commonest congenital cardiac anomaly in the adult affecting more than 30% of people and is the most common cause of cryptogenic cerebral infarction. What about prophylactic maze recommended by some in cases of ASD closure in adult patients which carries a 50% risk of AF [6]?

References