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Osler-Rendu-Weber syndrome: congenital arteriovenous intrapulmonary fistula treated using a percutaneous Amplatzer plug

V. CONTI[1], F. FIORUCCI[1], M. SERPILLI[1], A. PATRIZI[1], G. PAONE[1,4], P. NERI[1], G. GIANNUNZIO[1], M. GUZZO PIROZZO[3], C. FIORUCCI[2], G. LUCANTONI[1]

[1] C.U.B.E. Dipartimento di Scienze Cardiovascolari e Respiratorie, Università di Roma "La Sapienza", Ospedale "Carlo Forlanini", Rome (Italy)
[2] UOC di Radiologia, Ospedale di Bracciano, Rome (Italy)
[3] UOC di Medicina Generale, Fondazione Poliambulanza, Brescia (Italy)
[4] Fondazione Don Gnocchi, "Santa Maria della Pace", Rome (Italy)

Abstract. – Pulmonary arteriovenous fistulas (PAVFs) are rare vascular malformations (PAVMs) of the lung that could lead to severe hypoxiemia due to right-to-left intrapulmonary shunts. They may occur as isolated entities or associated with Osler-Rendu-Weber syndrome or hereditary haemorrhagic telangiectasia (HHT).

We report a case of a 70 years old woman with Rendu-Osler-Weber disease and a large arteriovenous malformation involving the left pulmonary artery.

We describe the successful transcatheter occlusion of the PAVF using an Amplatzer® vascular plug. This work is an attempt to focuse the attention on pulmonary arteriovenous malformations and on percutaneous treatment as an alternative to surgery, that consists of a conservative lung resection.

Key Words:

Pulmonary arteriovenous fistulas (PAVFs), Pulmonary arteriovenous malformations (PAVMs), Osler-Rendu-Weber syndrome, Amplatzer duct occluder (ADO).

Introduction

Pulmonary arteriovenous fistulas (PAVFs) are rare vascular malformations of the lung that could lead to severe hypoxiemia due to right-to-left intrapulmonary shunts. This condition is an abnormal passageway (fistula) between arterial and venous vessels, that occurs in lung vascular system. Patients with Osler-Rendu-Weber syndrome, also called hereditary hemorrhagic telangiectasia (HHT), frequently have abnormal blood vessels development in several sites[1,2].

This condition is slightly more common in women than in men. Many patients have no symptoms, some patients have difficulty breathing, shortness of breath with exertion, difficulty exercising, or bloody sputum (Table I).

We report a case of a patient who underwent a percutaneous interventional closure of a large intrapulmonary arteriovenous fistula using an Amplatzer duct occluder. This procedure was performed without complications and led to an improvement of arterial oxigen saturation and exercise tolerance. We believe it could be considered a valid alternative to open surgery.

Case Report

A 70-year-old female with a diagnosis of Osler-Rendu-Weber syndrome and a medical history of hypertension, myocardial ischemia and epilepsy, has been admitted to our Division because of epistaxis and severe dyspnea.

More than 15 years before, a lower lobe lesion in the left lung had been noted on a chest radiography. The first hypothesis was a lung cancer and the patient had undergone thoracotomy, resulted negative for malignant neoplasm. About 10 years ago, when the patient had started to experience fatigability, reduced exercise tolerance, exertional dyspnea and palpitations, she performed a chest tomography, that showed a large pulmonary arteriovenous fistula. She was not treated.

When admitted, the patient presented significant dyspnea. Physical examination showed dif-

Corresponding Author: Fabio Fiorucci, MD; e-mail: fabiofiorucci@tiscali.it

Table I. Pulmonary arteriovenous fistulas (PAVFs): signs and symptoms..

- The oxygen saturation level in the blood is low.
- There may be an abnormally high red blood cell count.
- A chest radiograph usually reveals the abnormal blood vessels.
- A chest computerized tomography (CT) (and RM) scan confirms the presence of the abnormal blood vessels.
- A pulmonary arteriogram is used to make a "road map" of the abnormal blood vessels before surgery or other treatments

fuse multiple scattered cutaneous teleangiectasic lesions especially localized in nasal and oral mucosa; central and peripheral cyanosis; digital clubbing. Cutaneous oxygen saturation values were between 86% and 88% at rest, falling to 84% with modest exertion. The hypoxemia was substantially refractory to supplemental oxygen: arterial oxygen partial pressure was 49.3 mmHg, breathing room air, and 51.1 mmHg with supplemental oxygen (8 liters at minute with 40 % Venturi Mask).

Laboratory tests showed a serum haemoglobin level of 10 gm/dl, red cells count of 3.75 millions/m and iron deficiency anaemia caused by epistaxis[3].

Electrocardiogram (ECG) showed a sinus rhythm, with signs of inferolateral myocardial infarction.

An echocardiogram revealed borderline left ventricular enlargement with lowered left ventricular function and good right ventricular function.

The finding on the chest radiography was a large circumscribed lobulated mass on the lower lobe of the left lung. Computerized Tomography (CT) underlined clearly the vascular nature of this lesion (Figures 1 A,B).

Magnetic Resonance Angiography confirmed the diagnosis and delineated a single isolated arteriovenous fistula draining from a large branch of the left pulmonary artery to the left superior pulmonary vein (Figures 2 A, B).

The patient has been treated by Amplatzer device, the therapeutic option for interatrial and interventricular defects, peripherical embolization of arteries and veins and pulmonary fistulas[4-7].

After treatment the patient has shown no longer dyspnea at rest, tolerance effort has increased, arterial oxygen partial pressure has been 61.3 mmHg breathing room air, and 75.1 mmHg with supplemental oxygen (4 liters at minute with 28% Venturi Mask).

These reports show that the shunt has been effectively corrected.

Discussion

Pulmonary arteriovenous fistulas (PAVFs) are abnormal direct communications between

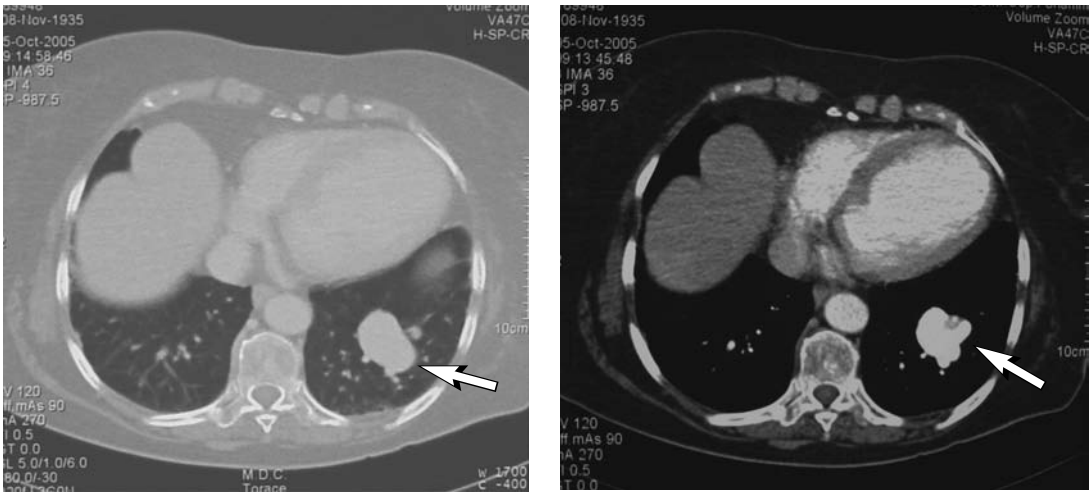

Figure 1. ***A, B***, Chest CT scan reveals a rounded lobulated mass (white arrow) in the left lower lobe, with intense contrast enhancement synchronous with the arterial vascular opacification.

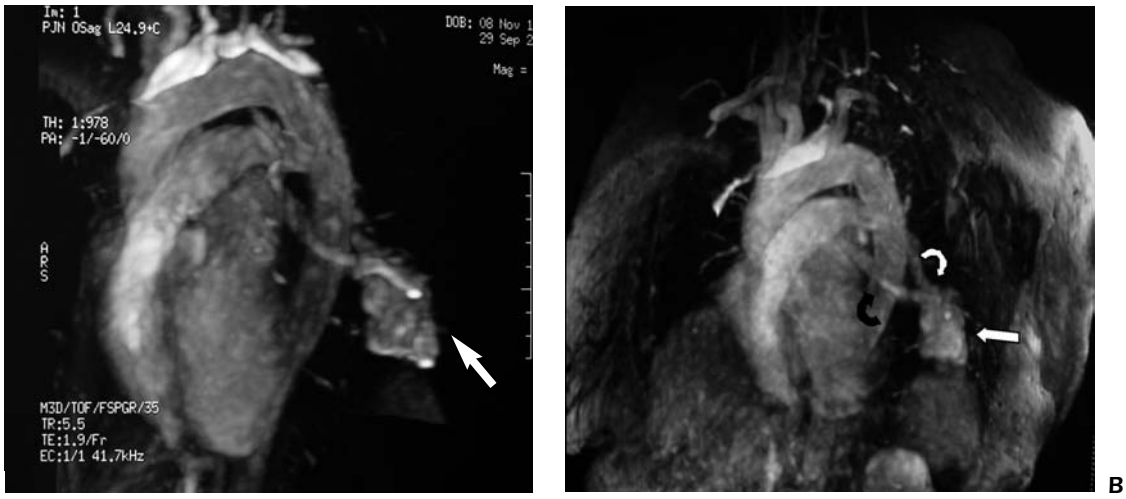

Figure 2. A, B, MR angiogram demonstrates the Arteriovenous pulmonary malformation (*white arrow*) in the left lower lobe, with a solitary feeding artery (*curve black arrow*) from a branch of the left pulmonary artery and a draining vein to the left superior pulmonary vein (*curve white arrow*).

branches of a pulmonary artery and pulmonary veins without an intervening capillary bed. More than 80% of PAVFs are congenital: most of these are associated with Osler-Rendu-Weber syndrome or HHT, an autosomal dominant disorder (one of the commonest monogenic diseases, with an incidence of one in 5-8000[1]) characterized by recurrent epistaxes, mucocutaneous telangiectasia, and visceral involvement, including arteriovenous communications that may develop in virtually any organ, especially in the lung[2,3]; overall, 5%-15% of the population with HHT have a PAVFs[2,3], usually in the lower lobes[3].

Secondary or acquired pulmonary arteriovenous malformations, although very rare, have been reported in literature, as a result of chest trauma, thoracic surgery, hepatic cirrhosis, metastatic carcinoma, infections, and systemic amyloidosis[2,4].

Symptoms are quite variable. PAVFs may often cause hypoxemia and dyspnoea due to right-to-left shunting, but remains frequently undiagnosed: the severity of dyspnoea is obviously related to the degree of hypoxemia and the magnitude of the shunt, but more than half of patients with PAVFs tolerate well hypoxemia and are relatively asymptomatic[4], unless the arterial oxygen pressure is less than 60 mmHg; the hypoxemia is often refractory to supplemental oxygen[2]. The classic triad of dyspnea on exertion, central or periferal cyanosis (or both) and clubbing is found in 30% of adults[4].

Typical clinical feature of patients with HHT is the presence of multiple telangiectasias at characteristic sites (lips, oral cavity, fingers, nose, but also in visceral sites, such as gastrointestinal tract, with recurrent epistaxis and melena, and consequent anaemia)[1].

In addition to lung, arteriovenous fistula may develop virtually in any organ, especially liver and brain: a history of neurological symptoms, headache, vertigo, paresis, numbness, paresthaesia, syncope, or confusion, can be found[2].

Complications of untreated PAVFs, even though asymptomatic, could be various: massive haemoptysis caused by intrabronchial rupture of a lesion, or haemothorax, due to rupture of a subpleural lesion, are rare, but potentially fatal[5]. The most common complications are related to central nervous system: catastrophic embolic cerebral events (cerebral abscess and embolic stroke), and transient ischemic attacks occur in sizeable number of patients (19-59%)[2,6]. The mechanism for these neurological events is paradoxical embolism across the PAVFs.

These complications can be limited if the condition is recognised and treated. The treatment of PAVFs has two crucial objectives: to eliminate or reduce the right-to-left shunt and to prevent and treat complications. Untreated lesions are associated with 11% mortality and 26% morbidity[4] and most patients with PAVFs should be treated.

The therapy of PAVFs is based on surgical treatment or transcatheter interventions[7,8]. Surgi-

cal treatment has significant morbidity and mortality and often requires removal of parts of the lung that are not involved with the PAVFs. Transcatheter interventions usually carry fewer risks that surgery and usually avoid the destruction of normal lung tissue.

The Amplatzer vascular plug device is a major therapeutic option to occlude pulmonary PAVFs. Several studies demonstrated excellent results of this intervention[9-11].

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