



Osler-Weber-Rendu Syndrome Complicated with Pulmonary Arteriovenous Malformation: A Case Report and Review of Literatures

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Osler-Weber-Rendu syndrome is a hereditary disease which is diagnosed by criteria of clinical symptoms and examinations. Here, we report a definite case of Osler-Weber-Rendu syndrome who had epistaxis, skin telangiectasia, and pulmonary arteriovenous malformation (PAVM). His initial clinical presentations were growing pulmonary nodule with cough and occasional chest pain. PAVM with rupture of aneurysm was diagnosed histologically after wedge resection of the nodule in his right lower lung.

Key words: Osler-Weber-Rendu syndrome, hereditary hemorrhagic telangiectasia, pulmonary arteriovenous malformation

INTRODUCTION

Pulmonary arteriovenous malformation (PAVM) is mostly a congenital and women predominant disorder. Though it is quite uncommon, recent studies have shown that PAVM could also be secondary to trauma, cirrhosis, mitral stenosis, actinomycosis, Fanconi's syndrome, and metastatic thyroid cancer. Clinical presentation varied from cases, but the most common symptom, epistaxis, may imply the possibility of the relationship with Osler-Weber-Rendu syndrome (also known as hereditary hemorrhagic telangiectasia [HHT]). According to previous literature, 70-90% of patients with PAVMs are associated with HHT,¹ and conversely, about 30% of patients with HHT have PAVMs.² Here, we report a case of HHT, who was diagnosed of PAVM in his right hemithorax, and was treated by thoracoscopic wedge resection.

CASE REPORT

A 57-year-old man with a history of chronic bronchitis under regular treatment, was referred to the chest surgery out-patient department for surgical consultation of the mass lesion in his right lower lung field. He had the habits of consumption of cigarettes (2 pack per day for more than 20 years), and liquors (200-500 ml of Kao-Liang) for more than 20 years. He was hospitalized about 10 years

ago because of pneumonia, and there was a nodular lesion found in his right lower lung field, which did not thought to be indicated for surgery. Three months ago, severe cough and occasional chest pain developed, which were refractory to medical treatment, thus he was hospitalized for further evaluation. He also had several episodes of spontaneous epistaxis in the past decades according to the medical record. Clubbing fingers, blood clots coating on the nasal mucosa, and small bruits auscultated from the right mid back were found during physical examination. His hemoglobin was 11.5 mg/dl, but alpha-fetal protein (5.14 mg/dl) and carcinoembryonic antigen (2.18 mg/dl) were within normal limits. Chest radiograph revealed a mass lesion about 2 cm in diameter superimposed on the right side 6th rib [Figures 1 and 2]. Another chest computed tomography (CT) scan was ordered [Figures 3 and 4], which disclosed a sharp-edged, approximately round-shaped mass lesion, with a feeding vessel supplied from the central lung. Echocardiography shows mild tricuspid regurgitation without significant cardiac function impairment, and pulmonary function test revealed normal results without patterns of obstructive lung disease. Video-assisted thoracoscopy found a capsulated lesion with pulsation over right lower lung field, and wedge resection was done smoothly. Grossly, the specimen showed a picture of abnormal vascular structures with a ruptured aneurysm, and the pathologist reported PAVM tissue under microscope [Figure 6].

Three months after surgery, the symptoms of cough had improved, and chest pain subsided. There were no more bruits or thrills auscultated from the chest, and clubbing fingers also disappeared. However, telangiectasia lesions were found on his right thigh medially and posteriorly. Further, brain CT study showed no vascular lesions.

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Wedge resection for treating PAVM

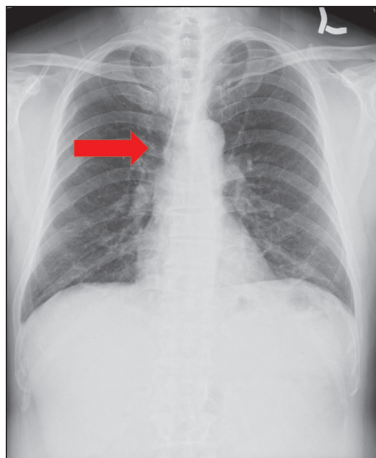


Figure 1. Initial chest radiograph obtained in 2009, showing a lung mass superimposed by the right sixth rib (arrow)

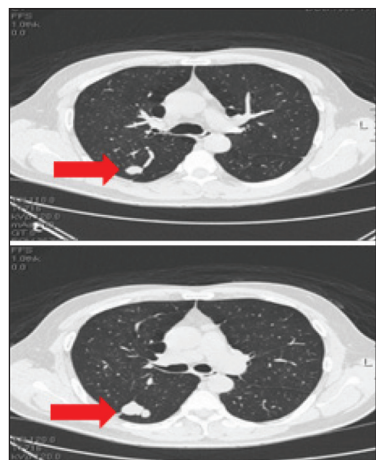


Figure 3. Chest computed tomography scan, axial view showed a nearly round-shaped mass lesion with feeding vessel

DISCUSSION

Pulmonary vascular disease includes embolism, congestion, hypertension, veno-occlusive disease, and AVM (PAVM). Aneurysm in pulmonary circulation is very rare because of relatively low arterial pressure. However, AVM provides a higher probability of the development of aneurysm in lung due to abnormal vascular wall content. In previous literature, pulmonary aneurysms were found centrally or together with AVM peripherally. The first case was reported in 1897, and more than 500 cases had been reported since then. Greater than 80% of PAVMs are congenital, and 80-90% of them are associated with Osler-Weber-Rendu syndrome (also known as hereditary HHT).¹ Conversely, 30-40% population of HHT has PAVM.

Hereditary HHT is an autosomal dominant disease, which may affect small portion of blood vessels at skin surface,

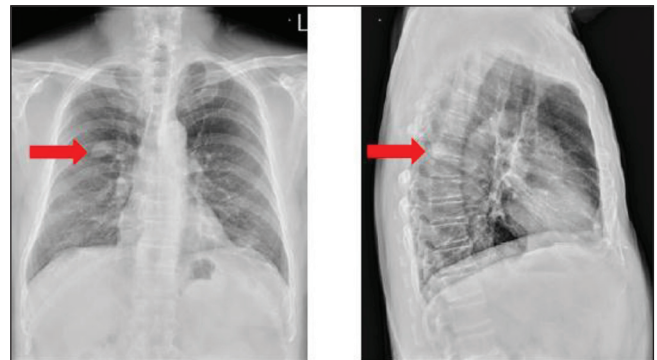


Figure 2. Chest radiograph in 2012, showing growth of right lung mass (arrow)

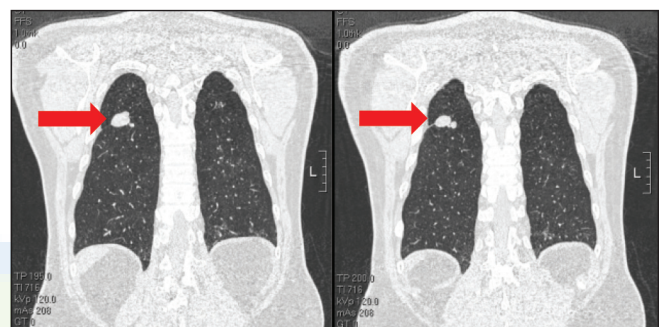


Figure 4. Chest computed tomography scan, coronary view

mucosa or rarely, organs. Most patients may experience nasal bleeding and skin telangiectasia lesions, and other symptoms may be associated with organs affected (e.g., headache or bruits in cerebral AVM, chest pain or dyspnea in PAVM, melena or hematochezia in gastrointestinal [GI] mucosal telangiectasia). The diagnosis criteria include: Nasal bleeding, multiple telangiectases over skin surface, internal lesions (GI tract, brain, spine, liver, lung), and a first-degree relative with HHT. Definite diagnosis may be made if 3 criteria are present, and a case that fulfills 2 criteria may be deemed as “possible.” If fewer than 2 criteria are present, it is an unlikely case of HHT.

Pulmonary arteriovenous malformations generally do not affect cardiac hemodynamics in contrast to systemic AVMs, however, degree of right-to-left shunt from pulmonary artery to vein determines the severity of clinical symptoms. Greater than 20% of blood shunting from right to left or a fall of more than 5 mg/dl of hemoglobin, may lead to obvious cyanosis, clubbing, and polycythemia.³ Diagnostic method vary from different capabilities and facilities in hospitals. Chest radiograph is essential, and may show abnormal findings in most of the patients with PAVM. A chest CT scan is often ordered to evaluate a lung tumor, in which associated blood vessels are identified under images with

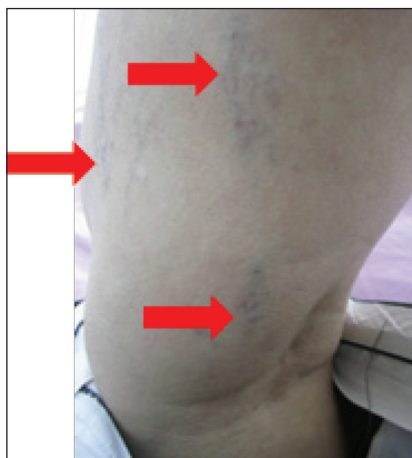


Figure 5. Multiple telangiectases on the right thigh, posteromedial aspect

and without contrast medium. If a PAVM lesion is suspected, then work up for right-to-left shunt is suggested. Contrast echocardiography is a highly sensitive diagnostic tool by injecting either agitated saline or dye from peripheral vein, and bubbles or dye would be observed in the subsequent cardiac echogram few seconds later, if right-to-left shunt exist. Pulmonary angiography is better in determining vasculature of individual PAVM, and carries the potential of treatment at the same time.

Treatment options include surgical and nonsurgical methods. All patients with associated symptoms or a lesion <2 cm, are indicated for intervention, either surgical or percutaneously.⁴ Another consensus suggested that PAVM with feeding vessels <2-3 ml requires adequate treatment.⁵ First surgical treatment, pneumonectomy, was successfully performed in 1942.⁶ Since then, patients with PAVM started to benefit from lobectomies (1950), and wedge resections (1959). Wedge resection of the lesion under thoracoscopy is the preferred technique if surgery is necessary.

The era of percutaneous therapy came after introducing in 1978 by Taylor *et al.*,⁷ and embolotherapy has become the mainstay of treatment options for PAVMs since then, using balloon or metallic coils. Case series reported by Pollak *et al.*¹ disclosed brilliant result of over 98% success rate of the procedure. Though there was no procedural mortality reported in the series, but central nervous system sequelae, including cerebral stroke and infection are another issue for whom receiving embolotherapy. Overall morbidity of untreated patients with PAVM is about 50%, but on the other hand, there is as low as only 3% in whom received treatment.^{4,8-12}

In our case, an AVM with aneurysm formation in his right lower lung incidentally, and he also had a history of

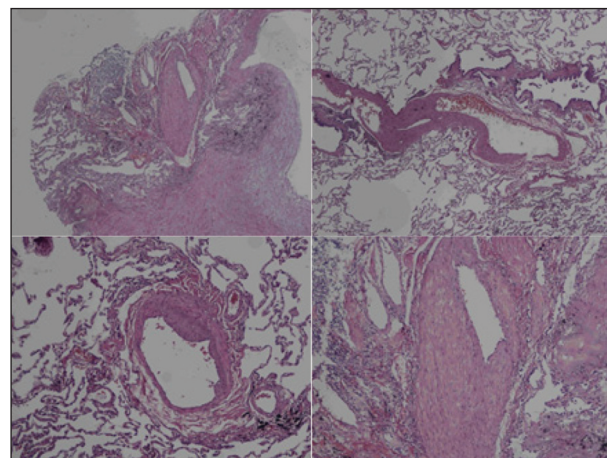


Figure 6. Histopathology: Arteriovenous malformation with aneurysm

several episodes of nasal bleeding, including this time of hospitalization. Furthermore, telangiectases were found on his right thigh [Figure 5]. Based on the above findings (fulfills three criteria of HHT), the man is a definite case of Osler-Weber-Rendu syndrome (HHT) with PAVM treated by wedge resection.

REFERENCES

1. Pollak JS, Saluja S, Thabet A, Henderson KJ, Denbow N, White RI Jr. Clinical and anatomic outcomes after embolotherapy of pulmonary arteriovenous malformations. *J Vasc Interv Radiol* 2006;17:35-44.
2. Guttmacher AE, Marchuk DA, White RI Jr. Hereditary hemorrhagic telangiectasia. *N Engl J Med* 1995;333:918-24.
3. Khurshid I, Downie GH. Pulmonary arteriovenous malformation. *Postgrad Med J* 2002;78:191-7.
4. Dines DE, Seward JB, Bernatz PE. Pulmonary arteriovenous fistulas. *Mayo Clin Proc* 1983;58:176-81.
5. Faughnan ME, Palda VA, Garcia-Tsao G, Geisthoff UW, McDonald J, Proctor DD, *et al.* International guidelines for the diagnosis and management of hereditary haemorrhagic telangiectasia. *J Med Genet* 2011;48:73-87.
6. Hepburn J, Dauphinee JA. Successful removal of hemangioma of lung followed by disappearance of polycythemia. *Am J Med Sci* 1942;204:681-7.
7. Taylor BG, Cockerill EM, Manfredi F, Klatte EC. Therapeutic embolization of the pulmonary artery in pulmonary arteriovenous fistula. *Am J Med* 1978;64:360-5.
8. Stringer CJ, Stanley AL, Bates RC, Summers JE. Pulmonary arteriovenous fistula. *Am J Surg* 1955;89:1054-80.

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9. Sluiter-Eringa H, Orie NG, Slutier HJ. Pulmonary arteriovenous fistula: Diagnosis and prognosis in non-complaint patients. *Am J Respir Crit Care Med* 1969;100:177-84.
10. Puskas JD, Allen MS, Moncure AC, Wain JC Jr, Hilgenberg AD, Wright C, *et al.* Pulmonary arteriovenous malformations: Therapeutic options. *Ann Thorac Surg* 1993;56:253-7.
11. Yater WM, Finnegan J, Giffin HM. Pulmonary arteriovenous fistula; review of the literature and report of two cases. *J Am Med Assoc* 1949;141:581-9.
12. Lee DW, White RI Jr, Egglin TK, Pollak JS, Fayad PB, Wirth JA, *et al.* Embolotherapy of large pulmonary arteriovenous malformations: Long-term results. *Ann Thorac Surg* 1997;64:930-9.

