CASE REPORT
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A Case of Embolization Seen in Pulmonary Arteriovenous Malformation in a Patient with Osler-Rendu-Weber Syndrome

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ABSTRACT

Osler-Rendu-Weber Syndrome or hereditary hemorrhagic telangiectasia (HHT) is characterized by telangiectasia of the skin and of the mucous membranes and intermittent bleeding from vascular abnormalities; in about 20% of the patients pulmonary arteriovenous malformation is present. Pulmonary arteriovenous malformation is a congenital anomaly in the lung which causes shunting of venous blood in the pulmonary artery to systemic circulation, resulting in cyanosis, polycythemia and clubbing. Recently we experienced a case of multiple pulmonary arteriovenous malformation associated with the telangiectatic change of the cerebral artery in a 16-year-old male patient, which was confirmed by pulmonary angiography. **(Korean Circulation J 2006;36:820-822)**

KEY WORDS : Osler-Rendu-Weber syndrome ; Arteriovenous malformation ; Telangiectasia, hereditary hemorrhagic.

Introduction

Osler-Rendu-Weber syndrome or hereditary hemorrhagic telangiectasia is an autosomal dominant disease, characterized by telangiectasia of the skin and of the mucous membranes with intermittent bleeding from vascular abnormalities. About 20% of patients displaying this syndrome are known to have a pulmonary arteriovenous malformation.[1] Pulmonary arteriovenous malformation is a rare congenital anomaly in the lung which causes shunting of venous blood in the pulmonary artery to systemic circulation.[2] The syndrome develops gradually without symptoms during infancy or childhood, and is usually identified incidentally through the 3rd and 4th decades of life. However, cyanosis, polycythemia and clubbing can develop and is characterized by the size of the lesion or severity of the malformation. Massive hemoptysis and paradoxical embolism may also occur.[3-5]

We present here a case of multiple pulmonary arteriovenous malformation associated with the telangiectatic change of the cerebral artery in a 16-year-old male patient with mild dyspnea on exertion, who was successfully treated with coil embolization. We also offer a brief review and discussion of the medical literature that has described this rare condition.

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Case

A 16-year-old man was admitted to our hospital complaining of mild dyspnea on exertion. He had an evaluation of the upper respiratory tract a year earlier in another hospital, including a chest X-ray, which revealed multiple nodules in both lungs. He was referred to our hospital and computed tomography of the chest showed the presence of multiple arteriovenous malformations in the lung: three in right lower lobe and one in left lower lobe. He was admitted to the hospital for definite treatment after a long period of observation. There was no other significant medical or family history related to the condition.

On admission, his blood pressure was 100/70 mm Hg, he had a heart rate of 94/min, and a respiratory rate of 22/min. On a chest examination, his breathing sound was clear and his heartbeat wa regular, without murmur nor thrill. There was no tenderness across the entire abdomen, and the liver and spleen were not palpated. An extremity examination revealed clubbing on both hands. The skin was unremarkable without any telangiectatic change.

Results of laboratory tests were as follows: CBC re-

vealed WBC 7400/mm^3, Hb 19.0 g/dL, platelets 305,000/mm^3. Urea and electrolytes, liver function test, urine and stool examination were normal. ABGA under room air showed a pH of 7.448, pCO_2 31.0 mm Hg, pO_2 67.6 mmHg, HCO_3^- 21.1 mmol/L, SaO_2 95.7% and did not show improvement with the administration of 2 L/min of oxygen: pO_2 66.3 mmHg, SaO_2 96.3%. Gastric fiberoscopy and nasal examination were unremarkable.

A chest X-ray demonstrated nodules in both lower lobes, with a connection to vascular marking (Fig. 1). A computed tomography of the chest revealed three nodules in the right lower lobe, with maximal size of 3.1 cm, and one nodule in left lower lobe (Fig. 2). A brain magnetic resonance image showed a small sized enhancing lesion at the left cingulated gyrus, suggesting a small vascular malformation including telangiectasia (Fig. 3). A pulmonary artery angiography revealed contrast pooling lesions with early venous drainage in both lungs, confirming the diagnosis of pulmonary arteriovenous malformation (Fig. 4).

We performed a therapeutic embolization to feeding arteries with two steel coils in the left lower lobe and with a total of nine coils in theright lower lobe. There was no residual contrast pooling lesion nor draining vein seen after treatment (Fig. 5). Post-embolization ABGA was improved: pH 7.425, pCO_2 42.4 mmHg, pO_2 97.8 mmHg, HCO_3^- 27.3 mmol/L, SaO_2 98.7%.

Discussion

Osler-Rendu-Weber syndrome was first described as a hereditary epistaxis by Babington in 1865. It was identified as one disease entity after descriptions by Rendu (1896), Osler (1901) and Weber (1907). In 1909, Hanes[6] coined the term 'hereditary hemorrhagic telangiectasia' in acknowledgement of the three features that defined the disorder. Goldman, et al.[7] reported

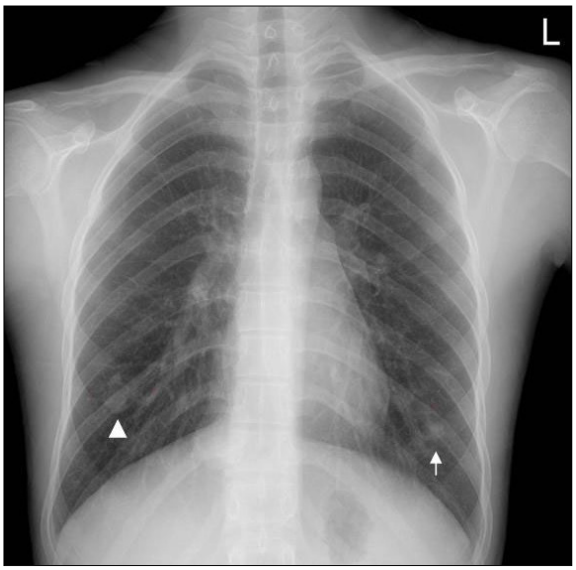

Fig. 1. Chest X-ray shows 3 cm sized well demarcated lobular mass in right lower lung field (arrow head) and 1 cm sized nodule in left lower lung field (arrow).

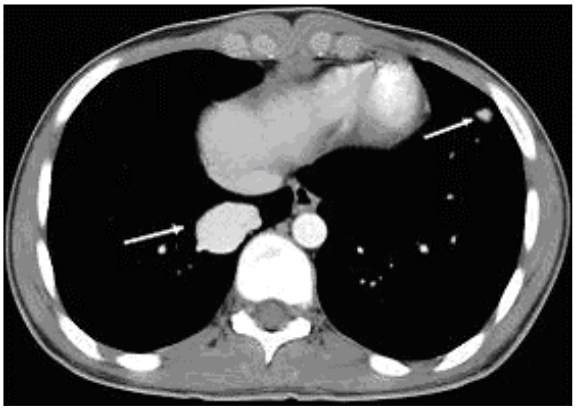

Fig. 2. Computed tomography of chest reveals markedly enhanced lobulating masses in both lung fields.

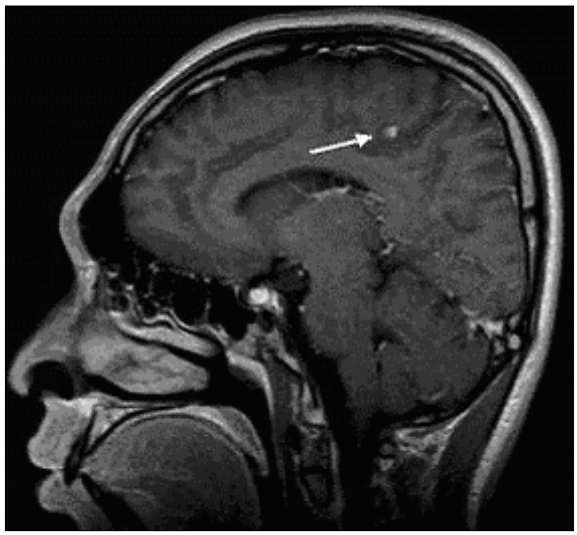

Fig. 3. A 5 mm sized enhancing lesion at the left cingulate gyrus area, suggesting small vascular anomaly or malformation including telangiectasia.

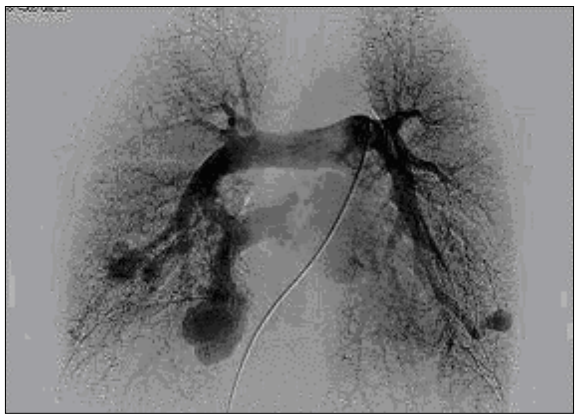

Fig. 4. Pulmonary arteriograms reveal pulmonary arteriovenous fistulas in both lungs. Contrast pooling lesions and early venous drainage patterns are noted.

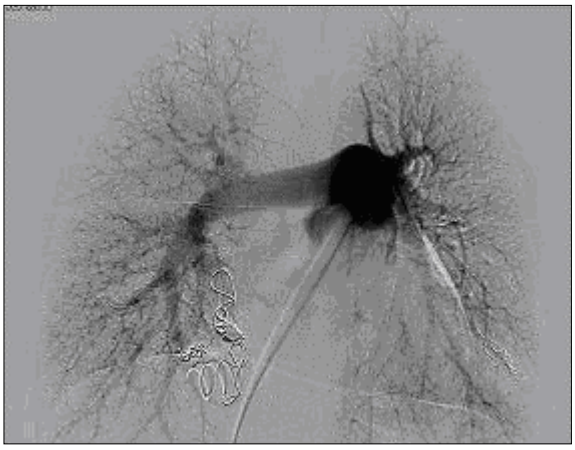

Fig. 5. Post-embolization with steel colis. Angiogram shows good embolization of pulmonary arteriovenous malformation without residual contrast pooling lesions.

that hereditary hemorrhagic telangiectasia is associated with 30-60% of patients with pulmonary arteriovenous malformation and appears to be a non sex-linked, autosomal dominant trait. The prevalence is about 1/8,000 to 1/100,000.[2] Telangiectasia of the skin and mucous membranes causes intermittent bleeding from vascular abnormalities, such as nasal bleeding, gastrointestinal bleeding and hematuria.[3]

Pulmonary arteriovenous malformation may be congenital or acquired. Congenital pulmonary arteriovenous malformation results from a persistent anomalous connection with the pulmonary artery and vein via a primitive splanchnic capillary bed.[5] It may be secondary to metastasis from thyroid cancer, pulmonary schistosomiasis, advanced liver disease, mitral stenosis and trauma.

The most common symptoms include hemoptysis and dyspnea. Cyanosis, clubbing and secondary polycythemia may occur according to the size of the lesion and the severity of the malformation.[5] Symptoms develop commonly after the 3rd decade of life. In hereditary hemorrhagic telangiectasia, extrapulmonary symptoms such as nasal bleeding, gastrointestinal bleeding and intracranial bleeding may occur.[3]

A diagnosis of pulmonary arteriovenous malformation is usually made by clinical manifestations, physical examination, chest X-ray, chest computed tomography and magnetic resonance imaging, contrast echocardiography and perfusion lung scintigraphy. A radiological exam will reveal well demarcated round or lobulated masses, occasionally connected to the communicating vessels. Pulmonary arteriography confirms the diagnosis, showing the number, size and contour of the lesions and vascular connections.[10] Magnetic resonance imaging is useful during follow-up.[10]

Congenital pulmonary arteriovenous malformation is a progressive disorder with a high complication rate at the onset of the symptoms. Because complications such as brain abscess, hemothorax, infective endocarditis and paradoxical embolism can be fatal if present, treatment is recommended whether the symptoms are present or not.[5]

Treatment consists of surgical resection and non-surgical intervention by therapeutic embolization through selective pulmonary artery catheterization. Surgical resection, the only treatment option available up to the 1970s, is limited to the patient with multiple lesions, pulmonary hypertension and severe obstructive lung disease. Therapeutic embolization, first introduced by Taylor and colleagues[9] in 1978, is effective for those complicated patients and yields a better result and lower complication rate. Surgical treatment should be reserved considering potential risks of the operation and decrease of the lung function.[8] While balloon and steel coils can be used for embolization, the use of a steel coil has advantages of easy insertion and permanent occlusion.[5] The most common complication of therapeutic embolization is the pulmonary infarction syndrome.

Prognosis is related to the association of the syndrome with cyanosis, hereditary hemorrhagic telangiectasia and complications. A good prognosis is seen following either surgical or non-surgical treatment.[1]

Our patient was asymptomatic except for mild dyspnea on exertion and had no familial history of the syndrome. However, the presence of multiple pulmonary arteriovenous malformation in association with cerebral arteriovenous malformation confirmed the diagnosis of Osler-Rendu-Weber syndrome. After therapeutic coil embolization, there was no residual contrast pooling lesions nor draining vessels with a subsequent improvement of ABGA, suggesting a good prognosis.

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