Large Aneurysms of the Ascending Aorta and Major Coronary Arteries in a Patient With Hereditary Hemorrhagic Telangiectasia

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We describe a 50-year-old man with a history of hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu disease) who presented with chest pain, atrial fibrillation, and congestive heart failure. Echocardiography revealed a large ascending aortic aneurysm accompanied by severe aortic regurgitation and giant coronary artery aneurysms involving the right, left main, left anterior descending, and circumflex coronary arteries. Coronary angiography clearly defined multiple aneurysms involving the aorta and coronary arteries. The patient underwent complex and successful surgical repair of the aneurysms. To our knowledge, this is the first reported case of extensive cardiac involvement in a patient with this uncommon genetic disorder.

Hereditary hemorrhagic telangiectasia (HHT) (Osler-Weber-Rendu disease) is an uncommon autosomal dominant disorder. Severe and recurrent epistaxis and skin mucosal telangiectases are universal features. More than 50% of patients with HHT also have pulmonary arteriovenous malformations. Some patients have brain abscesses or various forms of cerebrovascular accidents due to the underlying right-to-left shunt. Also, gastrointestinal bleeding and abnormal liver function may result from underlying arteriovenous malformations. Direct cardiac involvement is extremely rare.

REPORT OF A CASE

A 50-year-old man presented to our hospital with chest pain, palpitations, and substantial dyspnea on exertion. He had a lengthy history of HHT with multiple episodes of severe and life-threatening epistaxes that required numerous hospitalizations and interventions. He had chronic hypoxemia due to multiple pulmonary arteriovenous malformations that were treated with multiple coil embolization therapy. The patient had no traditional coronary risk factors such as hypercholesterolemia, hypertension, and diabetes mellitus. Although he was tall and thin and had long fingers, he had no history of Marfan syndrome. The patient had no history of ectopia lentis, pectus excavatum, arachnodactyly, or hypermobile joints. Echocardiography performed 7 years previously revealed aortic root enlargement to 4.1 cm with no evidence of aortic regurgitation. Three years previously, an ascending aortic aneurysm (6.0-cm diameter) with moderate aortic regurgitation was found, but the patient declined further surgical evaluation. On admission to our hospital, physical examination revealed a thin, chronically ill man with mild to moderate dyspnea at rest. His blood pressure level was 110/20 mm Hg, and his heart rate was 140 beats/min. Electrocardiography showed atrial fibrillation with rapid ventricular response. His oxygen saturation while breathing room air was 87%. Transthoracic echocardiography showed left ventricular enlargement, an ejection fraction of about 50% to 55%, a large ascending aortic aneurysm with a maximum diameter of 7.14 cm, severe aortic regurgitation, a giant right coronary artery aneurysm, and aneurysms of the left main, left anterior descending, and left circumflex coronary arteries visualized from parasternal views (Figure 1).

Selective coronary angiography revealed a giant right coronary artery aneurysm and extremely large aneurysms involving the entire left coronary artery system (Figure 2). The left coronary artery aneurysm showed filling defect and hypodensity, highly suggestive of thrombus formation. Several coronary stenoses appeared to be near the aneurysms. Supravalvar aortography confirmed a large ascending aortic aneurysm accompanied by severe aortic regurgitation.

The patient underwent successful aortic valve replacement, ascending aorta replacement with a Dacron graft, ligation of all the coronary artery aneurysms, and multiple saphenous venous bypass grafts to reestablish coronary flow. Surgical inspection confirmed the presence of multiple thrombi in the coronary artery aneurysms but revealed no evidence of atherosclerosis. The diameter of the right coronary artery aneurysm was up to 4 cm (larger than the...
normal-sized aorta), and the left coronary artery aneurysms were up to 3 cm. Pathological examination of the resected ascending aortic aneurysm showed patchy disruption of the elastic lamellar pattern of the media (Figure 3). There was no overt cystic medial necrosis, which can be seen in patients with Marfan syndrome or other connective tissue diseases. To our knowledge, this is the first reported case of extensive aneurysms involving the aorta and coronary arteries in a patient with HHT.

**DISCUSSION**

Guttmacher et al., in their review of HHT, summarized the main clinical manifestations of this disease. The usual sites involved include the nose, skin, lung, central nervous system, and gastrointestinal tract. The fundamental mechanism of this disease is related to the mutations of the endoglin leading to vascular dysplasia. Our case is unusual because the patient presented with a large ascending aortic aneurysm, severe aortic regurgitation, and giant coronary
artery aneurysms involving every major epicardial coronary artery, which we believe has not been reported previously. The prevalence of coronary artery aneurysm in one study was about 1.5% among 742 patients in whom angiography was performed. Coronary artery aneurysm is usually related to atherosclerosis, which our patient did not have. The chest pain our patient experienced probably was related to coronary artery thrombi, commonly present in coronary artery aneurysms. Physicians should consider the possibility of underlying HHT among the etiologies of coronary artery aneurysm (or notable ectasia) found on coronary angiography. Kurnik and Heymann were the first to report coronary artery ectasia in a patient with HHT.

There was a single reported case of coronary artery aneurysm associated with HHT in a 53-year-old woman. Koh et al described a single case of pulmonary artery aneurysm in a patient with HHT. Abnormalities of large systemic arteries associated with HHT were reported in 2 patients who had an extremely widened hepatic artery, a dilated aortic arch, and tortuous bronchial arteries. Trell et al reported that some patients with HHT had marfanoid habitus, a feature our patient did have, although he had no history of Marfan syndrome. This feature may signify the presence of underlying large systemic vessel disorders such as aneurysms involving the aorta and/or coronary arteries. Careful cardiovascular examination and noninvasive evaluation such as echocardiography can help identify aneurysms and/or valvular disease, facilitate definitive diagnosis and treatment, and prevent cardiovascular complications in patients with HHT.

REFERENCES