Acute Aortic Valvular Regurgitation Secondary to Avulsion of Aortic Valve Commissure in a Patient with Pseudoxanthoma Elasticum

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Abstract

A 68-year-old woman developed acute pulmonary edema due to severe acute aortic valvular regurgitation. At the time of emergency surgery, it turned out to result from spontaneous avulsion of the aortic valve commissure. Later, the patient was diagnosed to have pseudoxanthoma elasticum based on typical skin lesions. Connective tissue abnormalities associated with pseudoxanthoma elasticum might have contributed to the development of the avulsion of the aortic valve in this particular patient. (Internal Medicine 39: 940–942, 2000)

Key words: transesophageal echocardiography, connective tissue disorders, acute valvular regurgitation

Introduction

Although acute aortic valvular regurgitation is a relatively rare clinical condition, early correct diagnosis and adequate treatment including surgical intervention are crucial for survival of the patient. The most common cause of acute aortic valvular regurgitation is infective endocarditis followed by acute proximal aortic dissection and trauma (1). Rarely, spontaneous rupture of the aortic cusp may result in severe acute aortic valvular regurgitation in patients with connective tissue disease, such as Marfan syndrome (1). We describe a case of acute aortic valvular regurgitation resulting from spontaneous avulsion of the aortic valve commissure in a patient with pseudoxanthoma elasticum.

Case Report

A 68-year-old, previously healthy woman became suddenly dyspneic and was brought to the emergency room 10 hours after the onset of the symptoms. Initial vital signs included blood pressure of 150/70 mmHg, heart rate of 104 beats/min and respiratory rate of 30/min and temperature was 36.8 degrees Celsius. The patient was very sick and dyspneic. Skin was cold and clammy but there was no peripheral edema. Jugular venous pressure was not elevated and the carotid pulse showed a mild degree of bounding. Auscultation of the heart revealed Levine grade 4 (palpable thrill), monotonous holodiastolic murmur and grade 2 short systolic murmur at the left sternal border. Auscultation of the lung revealed wet inspiratory crackle with wheezing. A chest X-ray revealed severe pulmonary edema. A twelve-lead electrocardiogram showed high voltage in precordial leads and S-T depression of 0.1 mV in leads V1-V6, I and aVL. Arterial blood gas showed PH of 7.42 Torr, PCO2 of 28.8 Torr, PO2 of 85.9 Torr and HCO3 of 18.9 Torr (while breathing 10 l/min of oxygen by mask). Renal function was normal and there was no anemia. There was no elevation of cardiac enzymes.

A transthoracic echocardiogram revealed severe aortic valvular regurgitation. The left ventricle was not dilated and the wall motion was rather hyperdynamic. There was no intimal flap in the ascending aorta. The non-coronary cusp of the aortic valve showed possible prolapse. To evaluate more detailed anatomic information, a transesophageal echocardiography was performed. The noncoronary cusp was prolapsing down to the left ventricular outflow tract during early diastole resulting in loss of coaptation and severe regurgitation (Fig. 1A). There was no intimal flap in the sinus of Valsalva and ascending aorta. Because of hemodynamic instability, emergency surgical treatment was performed 16 hours after the onset of the symptoms. At the time of the surgery, cardiac surgeons noticed that the commissure between the noncoronary cusp and left coronary cusp was detached from the aortic wall [avulsion of the commissure (1)] and was prolapsing down to the left ventricular outflow tract (Fig. 1B). There was no perforation or rupture of the valves. The situation was overcome by suturing the detached commissure to the aortic wall at its original position.
Acute Aortic Regurgitation and Pseudoxanthoma Elasticum

Figure 1. A) A transesophageal echocardiogram (longitudinal view) showing detachment and prolapse of the noncoronary cusp (NCC) resulting in severe aortic valvular regurgitation. RCC: right coronary cusp, LA: left atrium, Ao: ascending aorta. B) An illustrative portrayal at the time of surgery depicting the avulsion of the commissure (as indicated by arrows) with prolapse of both noncoronary and left coronary cusps. LCC: left coronary cusp, NCC: noncoronary cusp (Modified from Carter JB, et al: Prolapse of semilunar cusps as causes of aortic insufficiency. Circulation 1971; 43: 922–932, with permission of the Lippincott Williams and Wilkins).

After the surgery, aortic regurgitation disappeared and congestive heart failure was successfully managed. Careful history taking and physical examination were repeated and revealed that one of her sons had had mitral valve plasty for severe mitral valve regurgitation secondary to mitral valve prolapse several years before. The patient’s height was 141 cm and weight was 48 kg. There was no deformity of the chest wall. High arched palate, joint laxity and arachnodactyly were not present. There were no angiod streaks in the retina, nor ectopia lentis, but blue sclera was present. There were orange peeling-like eruptions in the neck and chest and small papules in the axillar area. Skin folds were redundant in the axillar area. Biopsy of the skin lesion revealed disruption and fragmentation of the elastic fibers consistent with pseudoxanthoma elasticum (Fig. 2). Coronary angiography was performed and revealed no significant stenosis. Finally, this patient had congestive heart failure due to acute severe aortic valvular regurgitation, which resulted from the avulsion of the commissure between the noncoronary and left coronary cusps. The etiology of this rare condition appeared to be the connective tissue abnormality associated with pseudoxanthoma elasticum.

Discussion

Pseudoxanthoma elasticum is an inherited connective tissue disease that is associated with numerous heterogenous systemic manifestations (2). The most common cardiovascular involvement includes premature atherosclerosis leading to myocardial ischemia and intermittent claudication (3, 4). The pathogenesis of accelerated premature atherosclerosis is attrib-
uted to calcification of the internal elastic laminae of arteries (4). Sometimes the endocardium is also involved and results in restrictive cardiomyopathy (5). Reports of cardiac valvular involvement have been scarce and included mitral valve prolapse (6) and aortic regurgitation secondary to aneurysmal dilatation of the aortic annulus (7). The present case was unique because acute aortic valvular regurgitation due to the avulsion (detachment) of the commissure from the aortic wall occurred in a patient with biopsy proven pseudoxanthoma elasticum. The mechanism of valvular involvement has not been well defined. Lebwohl et al reported a high incidence of mitral valve prolapse in patients with pseudoxanthoma elasticum, and suggested that abnormal collagen or elastin might be the possible cause (6). On the contrary, Pyeritz et al had a negative impression regarding a close relation between mitral valve prolapse and pseudoxanthoma elasticum, except for type II pseudoxanthoma elasticum which shares the features of Marfan syndrome or Ehlers-Danlos syndrome type II (8). Heterogeneity of the clinical manifestations in one type of connective tissue disorders and some overlap of clinical manifestations in different types of connective tissue disorders are known to be common (9, 10). The diagnosis of pseudoxanthoma elasticum in the present case was confirmed from the typical biopsy findings. However, there was no significant atherosclerotic narrowing of the peripheral and coronary arteries. Further, the present case did not have Marfanoid features. The presence of blue sclera and a family history of mitral valve prolapse are more consistent with type II pseudoxanthoma elasticum, or the present case might have represented some “mixed” or “overlapping” type.

Another unique point in the present case was the auscultation findings. Usually the physical findings in acute valvular regurgitation are very different from those seen in chronic valvular diseases. The murmur of acute severe aortic valvular regurgitation may be small and confined to early diastole, or even absent because the totally incompetent valve allows equalization of aortic and left ventricular pressures rather early in diastole. There was a grade 4 diastolic murmur of aortic valvular regurgitation in this particular patient, and this was probably caused by the mechanical vibration of the detached commissure and related cusps, which prolapsed down to the left ventricular outflow tract.

Reference