

Surgical Treatment of Aortic Valve Papillary Fibroelastoma with Neurofibromatosis (von Recklinghausen's Disease)

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A 69-year-old man with a medical history of neurofibromatosis was transferred to our hospital for the treatment of an aortic valve mass. His workup identified an aortic valve papillary fibroelastoma based on the absence of signs of infection, thrombus in the left atrium, and valvular destruction or abnormal valvular function. A tumor was excised under cardiopulmonary bypass. The pathology of the mass was a papillary fibroelastoma. Neurofibromatosis with cardiac tumor is extremely rare. This is the first report of a case of papillary fibroelastoma associated with neurofibromatosis. (Ann Thorac Cardiovasc Surg 2010; 16: 213–215)

Key words: papillary fibroelastoma, neurofibromatosis

Introduction

Papillary fibroelastoma (PFE) is a rare heart tumor that occurs at a frequency of 2.5% to 7.9% among primary cardiac tumors.¹⁾ Its mechanism of development has not been determined. Neurofibromatosis is also a rare disease and is sometimes accompanied by cardiovascular lesions, but cardiac tumors have been reported only very rarely.²⁾ We encountered a unique case of neurofibromatosis with aortic valve papillary fibroelastoma.

Case report

The patient was a 69-year-old man with multiple subcutaneous nodules, *café au lait* spots, and deformity of the spine and thorax. Neurofibromatosis had been diagnosed, and a mass at the aortic valve was detected by echocardiography in a medical checkup 2 years earlier, but the mass was left untreated. A similar finding was detected

during a recent medical checkup, and the patient was referred to our hospital and admitted for closer examination.

He was short (height, 130.8 cm) and the thorax and spine were deformed. No cardiac or respiratory murmur was heard, but protruding neurofibroma and *café au lait* spots were present in several places over the body. Accurate assessment of the cardiothoracic ratio on chest X-ray was difficult because of marked deformity of the thorax. Severe scoliosis was noted on chest computed tomography (Fig. 1). No stenosis, dissection, or aneurysm formation was present in the aorta or peripheral arteries. Transthoracic echocardiography detected a 6.5 × 4.5 mm mobile mass attached to the right coronary cusp on the left ventricular side (Fig. 2). There was no functional abnormality of the valves or thrombus in the left atria. Coronary angiography was performed, but there were no findings of coronary artery stenosis, and aortography revealed no arterial regurgitation. PFE was diagnosed based on the absence of fever, inflammatory reactions, and abnormal valvular function, and we performed open heart surgery to prevent tumor embolism.

A median sternotomy was made during surgery. Extracorporeal circulation was initiated with blood withdrawn from the right atrium and returned to the ascending aorta. The aortic valve was tricuspid, and a 6 × 5 mm whitish pedicle mass was present in the belly of the right coronary cusp on the left ventricular side close to the left coronary cusp. The mass was mucoid with a papillary shape (Fig. 3).

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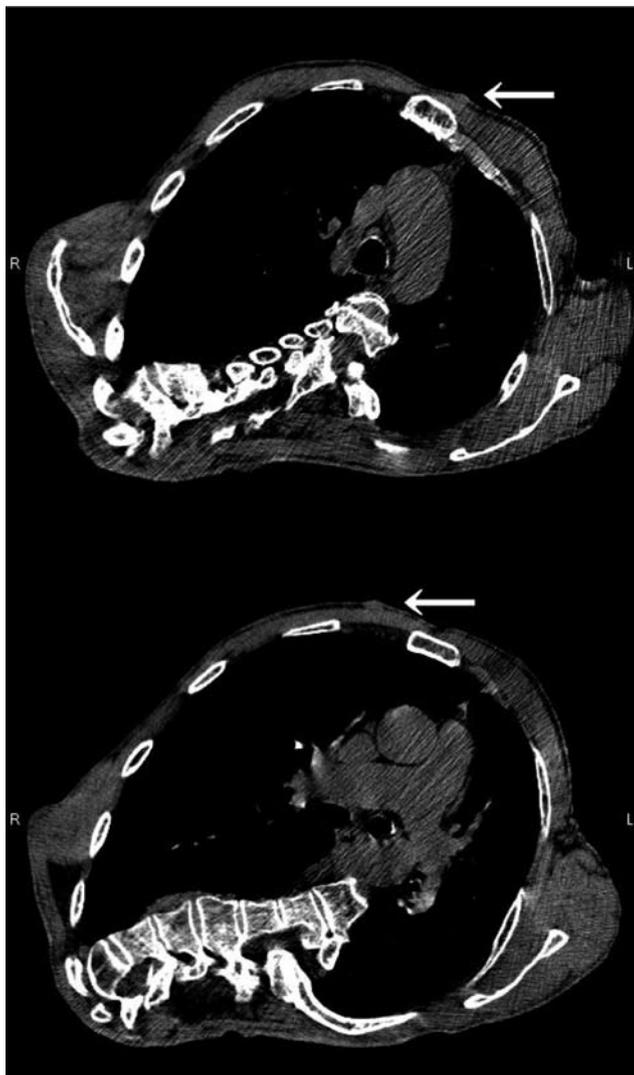


Fig. 1. Preoperative computed tomography demonstrated neurofibroma (arrow) and bone abnormalities, such as the thoracic deformity and scoliosis.

The tumor was excised by shaving the tunica externa of the right coronary cusp.

No residual tumor was detected on postoperative transthoracic echocardiography, and aortic valve function was good. On pathological examination, the mass was found to be a papillary tumor containing a collagen fiber core accompanied by myxomalike degeneration and was diagnosed as PFE (Fig. 4). The patient was discharged with no major complication. He is doing well and has no recurrence at 48 months after surgery.

Comment

Although PFE is a rare disease, it is well recognized, and

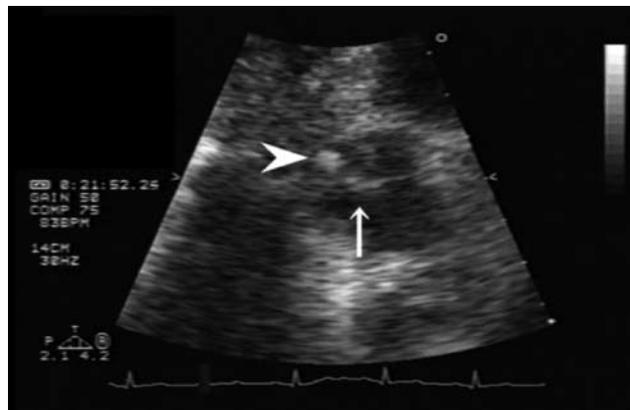


Fig. 2. Transthoracic echocardiography showed the mass with pendulum-like movement (arrowhead) attached to the right coronary cusp (arrow).

most surgeons are well informed about it. And as cardiac echoes are being performed more commonly, small PFEs are being recognized with increasing frequency.

Neurofibromatosis is also a rare disease. It is known as von Recklinghausen’s disease because it was described in detail by a German pathologist of that name, Friedrich Daniel von Recklinghausen, in 1882.³⁾ The incidence is about 0.025% to 0.033% (1/3000 to 1/4000), and the condition is caused by an aberration in chromosome 17.^{4,5)} The disease shows characteristic brown pigmented macules called *café au lait* spots and multiple subcutaneous nodules associated with neurofibroma. Furthermore, there may be changes in the bone of the spine, the four limbs, and thoracic vertebrae; pseudoarthrosis formation; and ophthalmological disorders, such as optic glioma and Lisch nodules.⁶⁾ Various cardiovascular lesions have also been reported, and many aspects of the disease have not been elucidated because of the diverse pathologies.

Malignant neoplasm is the most frequent cause of death, followed by cardiovascular lesions.⁴⁾ Complications resulting from cardiovascular lesions occur in 0.4% to 8.6% of cases of neurofibromatosis.^{2,5)} Diverse complications have been described, including coarctation and stenosis of arteries, aneurysm formation, intracardiac anomaly, valvular disease, and cardiomyopathy, but neurofibromatosis with cardiac tumor is extremely rare.²⁾ Neurofibroma or rhabdomyosarcoma have been described as complicating cardiac tumors in neurofibromatosis,^{2,7)} but to our knowledge, there is no report of benign tumor or PFE associated with this disease.

The development of PFE is generally thought to be associated with continuous turbulent blood flow into the endocardium, with resulting hyperplasia resulting from

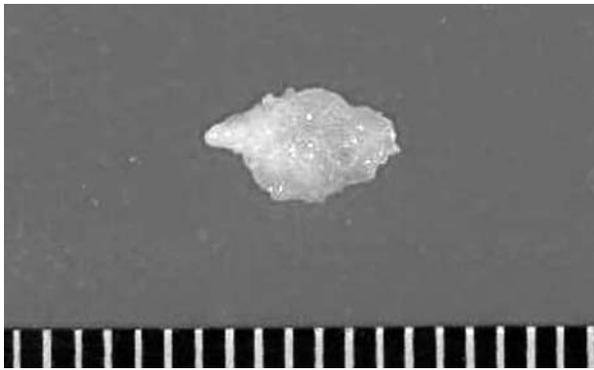


Fig. 3. The surgical specimen was soft and mixomatous.

endothelial cell impairment,¹⁾ and neurofibromatosis is associated with a histopathological picture of thickening of the tunica intima, thinning and disappearance of the tunica media because of atrophy, and vulnerability of the elastic laminae.⁵⁾ Our case may well be a completely incidental association of two relatively rare lesions. However, speculation from this basis suggests that valve tissue in the current patient may have become vulnerable, and PFE may have arisen as described above.

In summary, we performed tumorectomy for aortic valve PFE in a patient with neurofibromatosis and achieved a favorable outcome. Periodic follow-up is very important because a new cardiovascular lesion characteristic of neurofibromatosis may develop, including recurrence of PFE.

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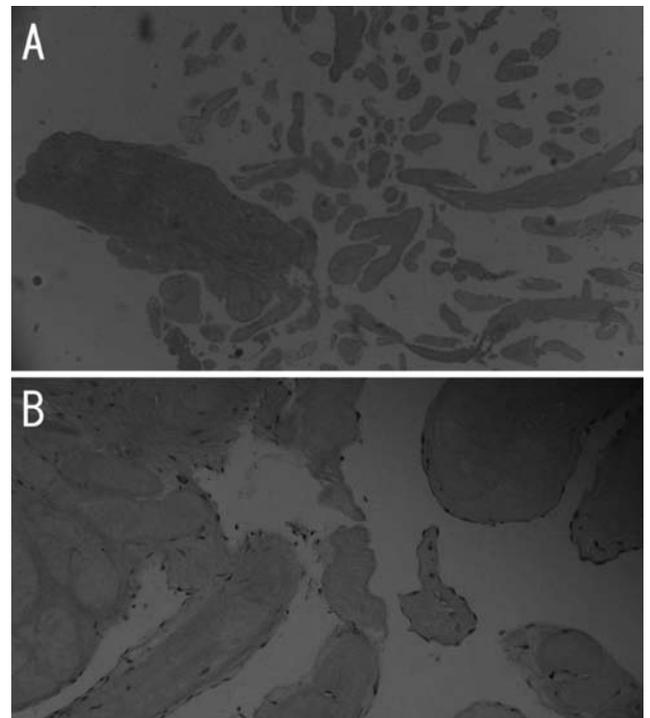


Fig. 4.

- (A) Pathology of the tumor showing papillary fronds. (Hematoxylin and eosin stain, $\times 40$ original magnification).
- (B) Histological picture of a few villi showing a collagen fiber core accompanied by myxoma-like degeneration. (Hematoxylin and eosin stains, $\times 100$ original magnification)

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