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CASE REPORT

Left ventricular myxoma with Carney complex

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Abstract

The left ventricle is a less frequent location of cardiac myxomas overall. Meanwhile, cardiac myxomas related to Carney complex (CNC), which is a multiple neoplasia syndrome involving cardiac, endocrine, neural, and cutaneous tumours, more frequently occur in the left ventricle compared with sporadic cardiac myxomas. Herein, we report a case of a 20-year-old woman with CNC who underwent complete surgical excision of a large and mobile left ventricular myxoma. In our case, echocardiography performed 4 years earlier was normal. This case highlights the importance of annual follow-up by echocardiography in patients with CNC, because early diagnosis of cardiac myxomas might improve their prognosis. Besides, we should bear in mind the possibility of CNC if the patients have cardiac myxoma in a cardiac chamber other than the left atrium at a younger age.

Keywords Carney complex; Cardiac myxoma; Tumour; Echocardiography

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Introduction

Cardiac myxoma is the most common primary heart tumour and located mainly in the left atrium. Meanwhile, cardiac myxoma with Carney complex (CNC), a rare multiple neoplasia syndrome involving cardiac, endocrine, neural, and cutaneous tumours,[1] can more frequently occur in the left ventricle.[2] We reported a young female patient with a left ventricular (LV) myxoma with CNC, which had a rapid growth rate and underwent complete surgical excision.

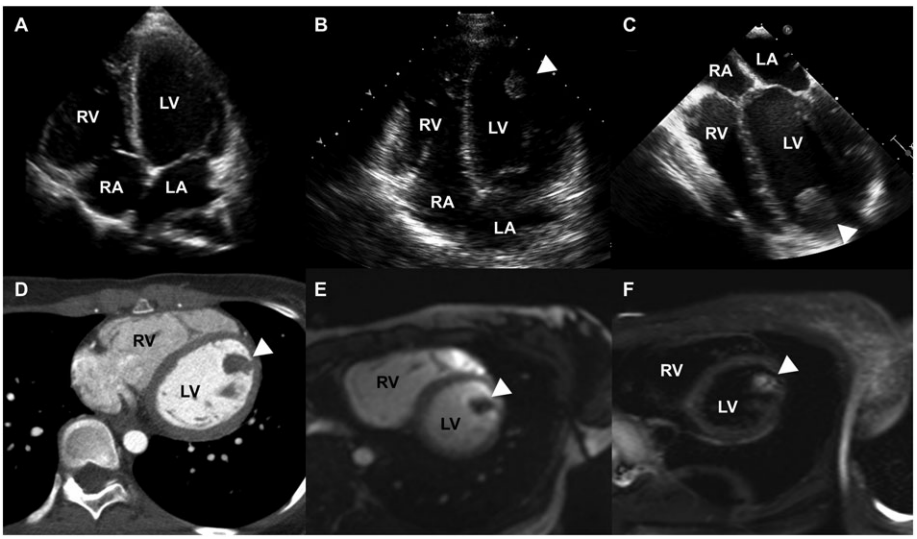
Case report

A 20-year-old woman was admitted to our cardiovascular centre for investigation of an LV tumour. She had been attending the outpatient clinic of the endocrine department of our centre for routine assessment after trans-sphenoidal resection of a pituitary tumour causing acromegaly at the age of 16. At that time, she was diagnosed with CNC by genetic screening, which revealed a somatic mutation of c.751_758del8 (p.S251LfsX16) in Exon 8 of the PRKAR1A gene on 17q24 in a pituitary tumour, given her endocrine tumour, spotty skin pigmentation with typical distribution, and family history (mother with Cushing syndrome and cardiac myxoma with CNC, elder sister with cutaneous myxoma with CNC, and grandmother with breast and cardiac tumours with CNC). Echocardiography performed at 16 years old showed no abnormal findings (*Figure 1A*).

At 20 years old, she was noted to have subcutaneous masses in the abdominal and sacral regions, and resection of these masses was planned. Echocardiography performed preoperatively happened to show a pedunculated, smooth-appearing mobile tumour of 22 × 14 mm attached to the LV apical–anterior wall (*Figure 1B*). The left ventricle had normal wall thickness and normal contractility. There was no LV outflow tract obstruction or valvular disease. She was referred to the cardiovascular department of our centre for further investigation.

On admission, she was asymptomatic, and physical examination findings were almost normal except for spotty

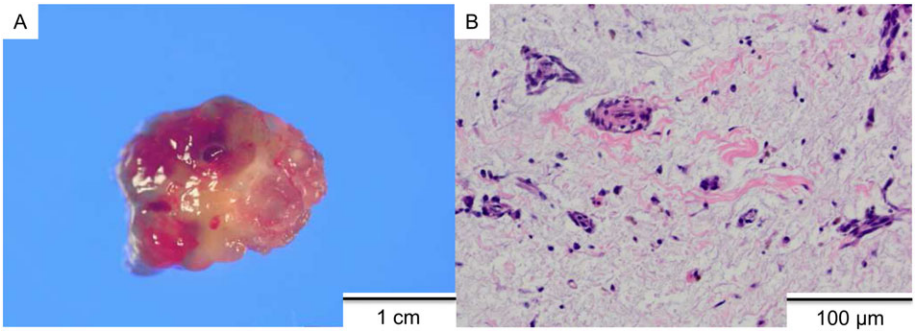
Figure 1 (A) Trans-thoracic echocardiography performed 4 years earlier. There was no abnormal finding. (B) Trans-thoracic echocardiography. A pedunculated, smooth-appearing mobile LV tumour was attached to the LV apical–anterior wall (white arrow). (C) Trans-oesophageal echocardiography. Only one tumour was found in the LV, and no tumour was found in other heart chambers (white arrow). (D) Contrast-enhanced computed tomography. Computed tomography showed that the LV tumour had a smooth margin, narrow stalk, and no contrast effect (white arrow). (E, F) Cardiac magnetic resonance imaging. The LV tumour was hypointense in T1-weighted images and hyperintense in T2-weighted images (white arrow). LA: left atrium, LV: left ventricle, RA: right atrium, RV: right ventricle.



skin pigmentation on her face and scoliosis of the spine. Results of blood tests including endocrine function, tumour markers, and interleukin 6 level were within the normal range. Trans-oesophageal echocardiography demonstrated that there was only one tumour in the left ventricle, with no other tumours found in either the left ventricle or other heart chambers (*Figure 1C*). Contrast-enhanced computed tomography (CT) demonstrated a pedunculated LV tumour attached to the LV anterior wall, with a narrow stalk without a contrast effect (*Figure 1D*). CT also showed no significant coronary stenosis and aneurysm or tumours of other organs. Cardiac magnetic resonance imaging revealed that the cardiac tumour was hypointense in T1-weighted images and hyperintense in T2-weighted images, which was compatible with cardiac myxoma (*Figure 1E*,*F*). Positron emission tomography–CT demonstrated no uptake of ^{18}F-fluorodeoxyglucose in the LV tumour and subcutaneous tumours.

Although the patient did not have any history of systemic embolization, she underwent urgent excision of the LV tumour via median sternotomy, considering that the tumour was mobile, had a narrow stalk, and was located in the left ventricle. The LV tumour was viewed through the mitral valve, and its stalk was located in the LV anterior wall. Thereafter, the tumour was resected en-bloc with a 2–3 mm margin of normal LV tissue from the stalk (*Figure 2A*), taking care of preventing migration of the tumour. Endocardium of the left ventricle and left atrium, including the mitral valve

Figure 2 (A) Macroscopic appearance of left ventricular tumour, showing a multinodular structure. (B) Microscopically, the tumour is composed of stellate and spindle cells in a background of myxoid extracellular matrix with no malignancy (haematoxylin and eosin staining).



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apparatus, was thoroughly investigated under cardiac arrest to confirm that the tumour solely existed in the left ventricle. As the tumour was resected en-bloc, irrigation of the LV chamber to wash out possible debris was not performed. Microscopic examination of the tumour revealed typical stellate cells in a background of a myxoid matrix with no malignancy, which indicated cardiac myxoma (*Figure 2B*). Tumour tissue was absent in the stump. The post-operative course was uneventful, and she was discharged 12 days after surgery.

Discussion

Carney complex is a multiple neoplasia syndrome involving cardiac, endocrine, neural, and cutaneous tumours with a variety of pigmented skin lesions.[1] It has an autosomal-dominant mode of inheritance. In our case, the patient and her family had a germline mutation in the *PRKAR1A* gene, which is a common germline mutation in patients with CNC. Her mother, elder sister, and grandmother were also diagnosed with CNC by genetic screening.

Carney complex characteristically occurs in young people (the mean age of disease onset is 26 years old), with a higher prevalence in females (62%).[2] Fifty-three per cent of patients with CNC have cardiac myxoma,[3] and nearly 7% of all cardiac myxomas are associated with CNC.[4] It is important to manage cardiac tumours in patients with CNC, since Stratakis *et al*. reported that the most common cause of death in patients with CNC was cardiac or heart related (57% of all-cause deaths, including cardiac myxoma, myxoma emboli, heart surgery complication, and probable cardiac arrhythmia). In patients with known CNC, annual echocardiography is recommended[5,6]; however, there are few reports demonstrating the developing course of cardiac myxoma in patients with CNC.[7] Annual echocardiography is sometimes forgotten in patients with known CNC, because that recommendation is based on anecdotal evidences. In our case, echocardiography performed 4 years earlier was normal, suggesting that tumour growth was relatively rapid. Thus, our case can emphasize the importance of annual echocardiographic examination in patients with CNC.

The left ventricle is a less frequent location of cardiac myxomas overall. Among all cardiac myxomas, left atrial cardiac myxomas are the most common (75%), followed by right atrial myxomas (15–20%), with left and right ventricular myxomas being the least common (3–4%).[8] On the other hand, cardiac myxomas related to CNC more frequently occur in the left ventricle, compared with sporadic myxomas. Edwards *et al*. reported that 64% of cardiac myxomas related to CNC occurred in the left atrium, 44% in the right atrium, 14% in the left ventricle, and 12% in the right ventricle.[2] According to previous reports, LV myxomas can cause systemic embolization, LV outflow tract obstruction, and systemic symptoms resulting from interleukin 6 production by tumour cells.[9] In our case, although the patient was asymptomatic, we decided to perform surgery, considering the large size and mobility of the tumour in the left ventricle. As a result, complete surgical excision of the tumour was accomplished with no complication. In our case, the cardiac tumour was detected after the diagnosis of CNC. On the other hand, some patients with CNC are diagnosed following detection of a cardiac tumour as the initial presentation. If the patient has a cardiac myxoma, especially in a cardiac chamber other than the left atrium at a younger age, we should bear in mind the possibility of CNC.

Conflict of interest

None declared.

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