First case of cardiac amyloidosis presenting as right atrial mass

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Introduction

Intracardiac masses can be encountered during the evaluation of patients with cardiac symptoms; myxoma, primary and metastatic neoplasms, thrombus, and vegetation should be considered in the differential diagnosis in such cases. Intracardiac masses can cause symptoms such as embolization, heart failure, valvular regurgitation, arrhythmia, and pericardial effusion. Although some of the symptoms may overlap, cardiac amyloidosis (CA) is not an expected diagnosis when investigating an intracavitary mass. Different from the previously reported cases, we report the case of a patient with CA who presented with a unique right atrial mass.

Case Report

A 62-year-old man with diabetes mellitus was admitted to our clinic with lower limb edema, shortness of breath, and fatigue. His electrocardiogram (ECG) and chest radiogram did not show any specific signs (Fig. 1a, 1b); thus, transthoracic echocardiography (TTE) was performed, and it showed minimal pericardial effusion, mild tricuspid regurgitation, and paradoxical movement of the interventricular septum. His medical history included undergoing right heart catheterization and pericardiocentesis thrice, suggesting constrictive pericarditis (CP). His blood tests showed no remarkable abnormality except mild hypochromic microcytic anemia (12.4 g/dL). Subsequently, right and left heart catheterizations were planned to resolve his symptoms. Right heart catheterization confirmed the diagnosis of CP, for which the patient was followed up with medical therapy. Furthermore, a coronary stent was implanted in the left anterior descending coronary artery because of significant stenosis. Seven months later, the patient was admitted to our hospital due to atrial fibrillation with low ventricular rate accompanied by dizziness. Surprisingly, his TTE showed a new intracardiac mass attached to the bottom of the right atrium. Transesophageal echocardiography (TEE) was performed, but it could not detailing the mass (Videos 1-6). Thereafter, cardiac magnetic

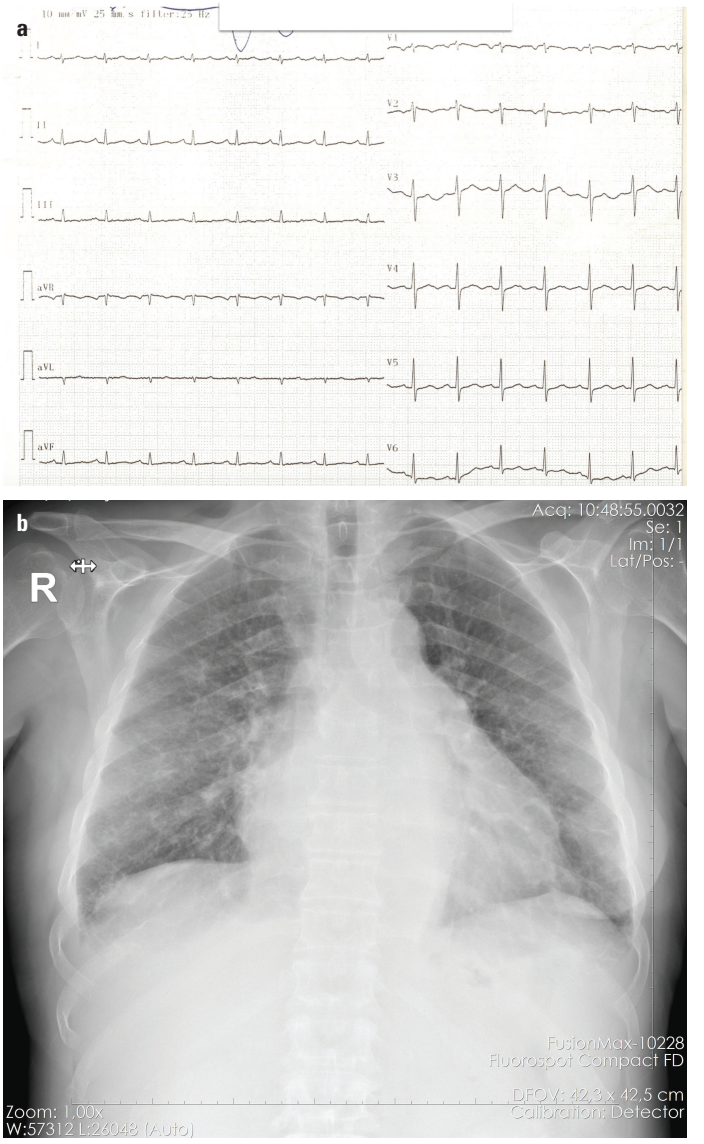

Figure 1. (a) The patient's electrocardiogram showed sinus rhythm without low-voltage QRS or pseudoinfarction pattern. (b) The chest radiogram displayed mildly increased cardiothoracic ratio

resonance (CMR) imaging was performed. It demonstrated an intracardiac mass with extensive gadolinium enhancement, suggesting an infiltrative disease, but did not indicate a specific diagnosis (Fig. 2a-2c). Thus, we performed a TEE-guided percutaneous endomyocardial biopsy targeting the masses under fluoroscopy and taking samples from both the masses separately. Microscopic examination of six different biopsy samples led to a diagnosis of CA due to apple-green birefringence with Congo red stain under polarized light microscopy. Pathological diagnosis was AA-negative amyloid. Hematological test and bone marrow biopsy results led to the exclusion of the diagnosis of AL amyloid. The patient discontinued visiting the outpatient clinic during follow-up when the amyloidosis subtype was being investigated.

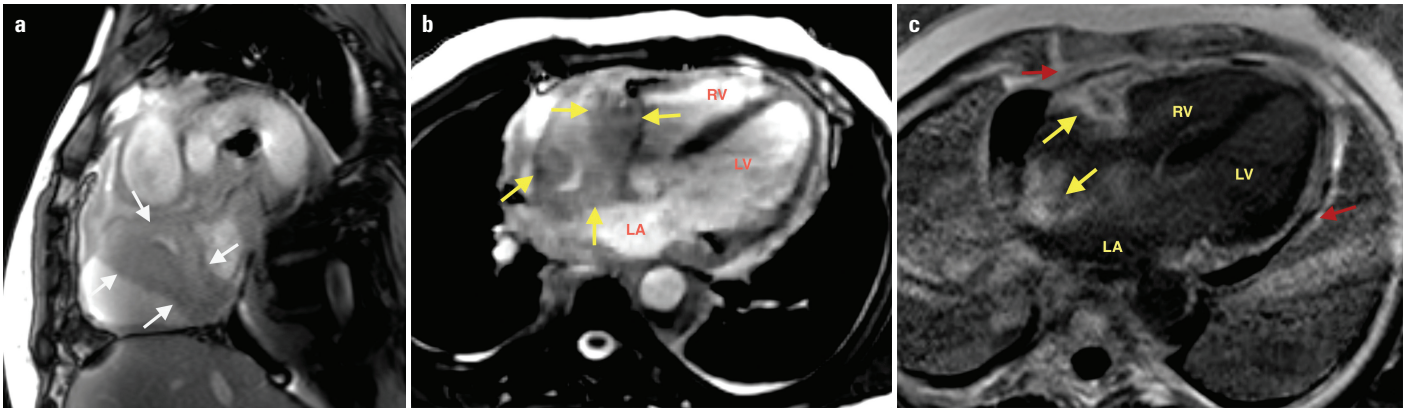

Figure 2. (a) Short axis GE (gradient echo) T2-weighted image shows a peripheral mass measuring 2–2.6 mm in thickness in the right atrium and atrial septum (white arrows). (b) Four-chamber GE T2-weighted image shows a peripheral mass comprising two separate parts (yellow arrows). Pericardial fluid and pericardial thickening were also observed. (c) Late gadolinium contrast-enhanced four-chamber image shows significant enhancement in the mass (yellow arrows) and both layers of the pericardium (red arrows)
LA - left atrium, LV - left ventricle, RV - right ventricle

Discussion

To the best of our knowledge, this is the first case of CA presenting as a solitary mass. The differential diagnosis of a right atrial mass should include vegetation, thrombus, myxoma, and cardiac neoplasms. The most common tumors of the right atrium are myxomas; they usually present on a stalk or attached to the interatrial septum (1). If the mass is layered against the wall of the atrium, a thrombus should be considered, especially in patients with central venous catheters or increased atrial size. If the mass invades the myocardium, initial diagnosis is a cardiac tumor. Vegetations are commonly located on the heart valves accompanying an infectious condition.

CA, which is usually not considered in the differential diagnosis of an intracavitary mass, can exhibit pericardial effusion, myocardial hypertrophy, granular sparkling pattern, or atrial dilatation on TTE images. Furthermore, diffuse subendocardial gadolinium enhancement, which appears in the late stages of the disease, can be seen on CMR imaging (2).

Patients with CA often present with shortness of breath, peripheral edema, and fatigue (2). General low-voltage or pseudoinfarction pattern on ECG is a commonly encountered sign that may not be detected in all cases, as in our patient (3). CA can be diagnosed via endomyocardial biopsy or imaging evidence of the heart signing to amyloidosis in the presence of histologic evidence of amyloid on another tissue (4).

CA occurs as a result of primarily three types of amyloidoses (5). AL amyloid is caused by monoclonal gammopathy; hereditary form appears as a result of a mutation in transthyretin protein, and senile amyloid is caused by wild-type transthyretin (6). Identification of the subtype is essential for deciding the therapeutic approach. The management of AL amyloidosis includes chemotherapy and stem cell transplantation; the hereditary form can be cured with liver transplantation, and the treatment of senile amyloidosis comprises supportive care (2).

Conclusion

The diagnosis of CA can be overlooked because of its non-specific clinical presentation. Clinical suspicion is crucial for early recognition, as clinical outcomes are poor with late diagnosis. Therefore, physicians should be familiar with atypical presentations of CA such as an intracardiac mass.

Informed consent: An informed consent was obtained from the patient by phone.

Video 1. 0-degree four-chamber view showing no myocardial hypertrophy
LA - left atrium, LV - left ventricle, RA - right atrium, RV - right ventricle

Video 2. 0-degree deformed view displaying a mass attached to the bottom of the right atrium and localized pericardial effusion near the right atrium
M - mass, PE - pericardial effusion, RA - right atrium, RV - right ventricle

Video 3. 66-degree view exhibiting two separate mass in the right atrium
M - Mass, RA - right atrium

Video 4. 115-degree bicaval view exhibiting the mass comprising two separate parts
M - Mass, RA - right atrium, SVC - superior vena cava

Video 5. 115-degree bicaval view showing intermittent occlusion of the entrance of superior vena cava
M - Mass, RA - right atrium, SVC - superior vena cava

Video 6. 123-degree bicaval view showing blocked blood flow from the superior vena cava
LA - left atrium, M - mass

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