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Cardiac amyloidosis

SHORT CASE REPORT

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Background

Cardiac amyloidosis is a rare condition often recognised as restrictive cardiomyopathy caused by extracellular accumulation of proteins in the myocardium. A more seldom form is amyloidoma, a solitary, localised tumour-like deposition of amyloid.

Case presentation

A man in his forties presented with lethargy, chest discomfort and fever over the preceding three weeks. Transoesophageal echocardiography (TEE) revealed an intracardiac tumour causing severe obstruction and secondary increased diastolic pressure gradient between the left atrium and ventricle. Emergency surgery revealed large tumour masses in both atria, predominantly on the left side. The patient died perioperatively within 24 hours of admission to the local hospital. Histopathological examination confirmed the diagnosis of amyloidoma.

Interpretation

Cardiac amyloidoma is an extremely rare finding that can lead to severe haemodynamic compromise. This case highlights the importance of considering cardiac tumours in young adults with atypical cardiac symptoms and urgent investigation with TEE.

Tumour-like amyloid deposits in the heart are exceedingly rare. We present a case report of an amyloid cardiac tumour with severe haemodynamic compromise.

A previously healthy man in his forties was admitted to a local hospital after three weeks of pronounced fatigue and reduced exercise tolerance. He had mostly just been lying asleep on the sofa, but reported intermittent inspiratory, stabbing and constrictive chest pain that worsened with exertion.

On admission to hospital, he was alert and oriented and did not appear to be in pain. His temperature was 38.5 °C, blood pressure 105/81 mmHg, oxygen saturation 98 % and sinus tachycardia was 112 beats per minute. Electrocardiogram (ECG) showed no signs of ischemia or conduction abnormalities. Cardiac auscultation revealed no murmurs, and additional investigations, including ECG, chest radiography and handheld cardiac ultrasound, showed no obvious pathology. Blood tests revealed mildly elevated

inflammatory markers: CRP 16 mg/L (< 5), leukocytes 15 × 10^9/L (3.5–10 × 10^9/L), and markedly elevated troponin I at 308 ng/L (< 34). D-dimer was normal (< 0.5 mg/L), and other laboratory parameters were unremarkable.

The patient was transferred to the cardiology department for further diagnostic evaluation and management. Over the following hours, he developed hypotension, which was managed with approximately 2500 mL of intravenous Ringer's acetate. Due to persistent fever and tachycardia, possible sepsis was considered. Blood cultures were therefore obtained, and broad-spectrum intravenous antibiotics were initiated: penicillin 3 g × 4 and gentamicin 400 mg × 1. The patient subsequently developed respiratory failure, with oxygen saturation dropping to 89 %. Pulmonary oedema was confirmed on bedside chest radiography, and continuous positive airway pressure (CPAP) therapy was initiated.

A comprehensive transthoracic echocardiogram (TTE) performed approximately 14 hours after admission (Figure 1a) showed a hyperechogenic structure in the mitral annulus bridging the left atrium and ventricle, with markedly elevated diastolic pressure gradients. Systolic left ventricular function was normal, and all four cardiac chambers were of normal size, with no signs of pulmonary hypertension.

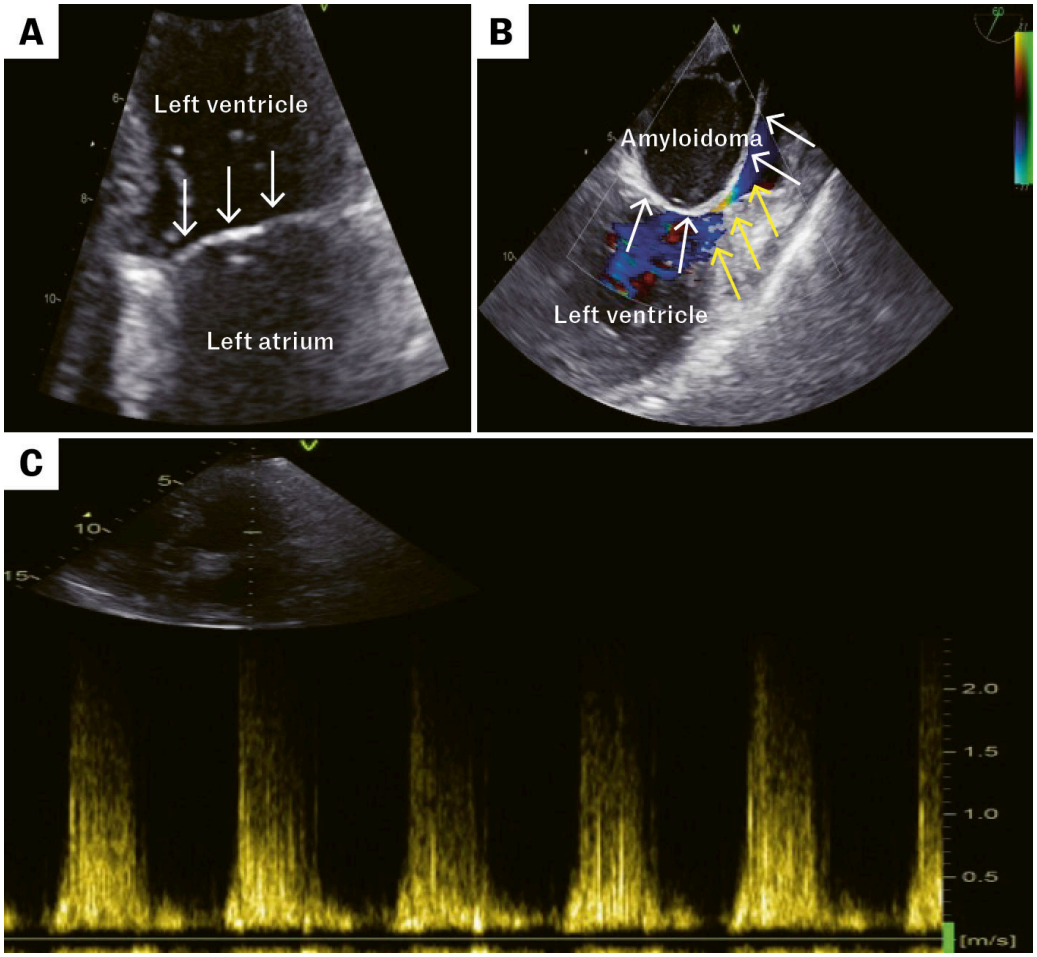

Figure 1 Echocardiography. a) Transthoracic echocardiographic image showing a bridge-like structure (white arrows) near the mitral valve. b) Transoesophageal image with colour Doppler demonstrating blood flow (yellow arrows) adjacent to the atrial mass. c) Transthoracic continuous-wave Doppler measurement across the mitral valve outflow, showing high velocities and elevated pressure gradients.

The patient's condition deteriorated, with blood pressure falling below 90/60 mmHg (mean arterial pressure 65 mmHg), reaching a nadir of 74/49 mmHg (57 mmHg). Progressive sinus tachycardia (120–130 beats/min) and anuria were also observed. A norepinephrine infusion was therefore initiated, titrated up to 0.3 µg/kg/min. CPAP therapy failed to achieve respiratory stabilisation. Approximately 18 hours after admission, the patient was sedated with fentanyl, midazolam and ketamine, intubated and mechanically ventilated. He was transferred to the intensive care unit (ICU).

Rapid transoesophageal echocardiography (TEE) was performed (Figure 1b), revealing a bridge-like hyperechogenic structure in the mitral annulus forming the outer layer of a mass that occupied over 95 % of the left atrial cavity. The mass left little space for blood to flow from the pulmonary veins through the left atrium into the left ventricle (Figure 1b). The diastolic pressure gradient between the left atrium and ventricle was markedly elevated, with a mean gradient of 18 mmHg (normal 0–5 mmHg) (Figure 1c).

A cardiac tumour was suspected, and the patient was transported by air ambulance to the university hospital for emergency surgery. Large tumour masses were found in both atria, predominantly on the left side. Unfortunately, the patient's condition deteriorated further, and he died during surgery approximately one day after admission to the local hospital.

The myocardial resection containing the tumour was sent to the pathology department, where special staining with Congo red demonstrated apple-green birefringence. Immunohistochemistry for amyloid P was positive, and transmission electron microscopy revealed homogeneous deposition of amyloid proteins, supporting the diagnosis of amyloidoma (Figure 2). Mass spectrometry identified predominantly proteins of the atrial natriuretic peptide type. Autopsy revealed no amyloid deposits in other organs, but signs of pulmonary oedema were present, and the cause of death was considered to be heart failure secondary to atrial amyloidoma.

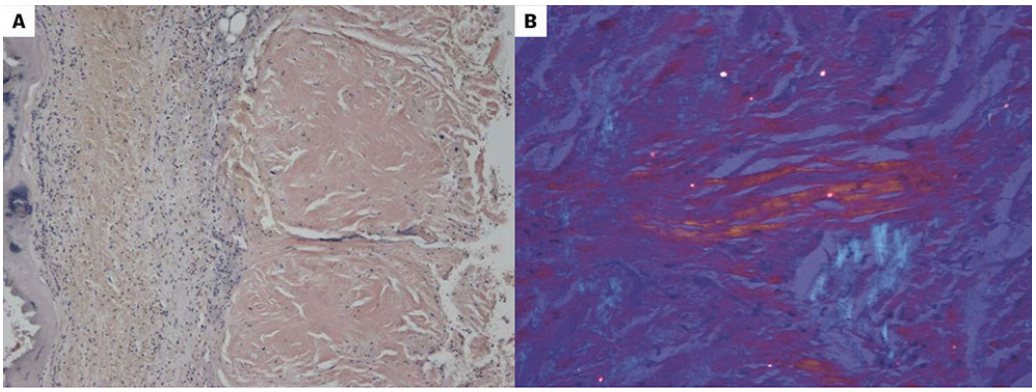

Figure 2 a) Congo red–stained tumour section (20× magnification) showing myocardium with condensed eosinophilic cytoplasmic deposits and focal calcifications. b) Red-yellow-greenish fluorescent cytoplasmic deposits (20× magnification).

Discussion

The cardiac mass was found to be an amyloidoma, a tumour-like amyloid deposit.

Many conditions can present as cardiac masses, including vegetations, thrombi, sarcoid deposits and myocardial abscesses (1). In general, cardiac metastases are far more common than primary cardiac tumours, with a reported ratio of approximately 20:1. Over 90 % of primary cardiac tumours are benign, with myxoma being the most common and predominantly located in the left atrium. These lesions can lead to obstruction (2). Malignant primary cardiac tumours are mostly sarcomas and lymphomas (2). Cardiac amyloidosis typically presents as restrictive cardiomyopathy caused by extracellular deposition of proteins in the myocardium (2). This form of amyloidosis may be primary, associated with monoclonal plasma cell disorders, or secondary, for example related to hereditary factors or chronic inflammatory conditions. Amyloidoma is a rare manifestation of amyloidosis, which itself encompasses a spectrum of deposition disorders in which nine of more than 30 different proteins can form amyloid fibrils deposited in the heart (2). Amyloidomas can occur in various anatomical sites, including the skin, chest wall and brain (3), but there are few reported cases of cardiac amyloidoma (4–7). In our patient, the deposits were predominantly composed of atrial natriuretic peptide-type amyloid, a pattern seen in patients with isolated atrial amyloidosis, where the amyloid is confined to the atria (8).

During surgery, large tumour masses were observed in both atria. However, the cardiac amyloidoma proved difficult to detect by TTE due to limited acoustic windows. Although Doppler assessment on TTE suggested obstruction with impaired left ventricular filling, resembling severe functional mitral stenosis, TEE was needed to identify the atrial tumour. Although rarely employed as an initial diagnostic tool in the emergency setting, TEE could have allowed earlier detection of the mass. Chest CT could also have been considered at admission, before clinical decompensation made such imaging more challenging.

It is unclear whether more conservative treatment with inotropes, vasopressors and fluids could have reduced cardiac strain and potentially prevented the rapid progression to heart failure and pulmonary oedema. However, the patient presented with a diagnostically unclear condition and a clinical picture resembling septic shock, in which haemodynamic support is often necessary to maintain organ perfusion. The patient's young age and good physical condition may have helped compensate for the initial hemodynamic effects.

Our aim with this case report is to document an exceedingly rare finding. Only a few cases of intracardiac amyloidoma have been reported internationally (4–7). We also wish to emphasise the importance of high-quality echocardiographic assessment and to demonstrate the potential severe hemodynamic effects of an obstructive cardiac mass, regardless of its aetiology.

This is clinically relevant because, although cardiac amyloidoma is extremely rare, the symptomatology may be applicable to a broader group of patients regularly encountered in clinical practice.

The authors were unsuccessful in their attempts to contact the patient's next of kin. The article has been anonymised to the greatest extent possible and is being published without consent, as the content is considered to be of significant public interest and there is no reason to believe that the patient or his next of kin would have objected.

The article has been peer-reviewed.

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