CLINICAL IMAGE

Cardiac amyloidosis: myocardial biopsy as a tool in chemotherapy implementation and sudden cardiac death prevention

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A 63-year-old woman with a short history of orthostatic fainting and heart palpitations was admitted to the hospital for further diagnostic workup. Preliminary diagnostic tests, including transthoracic echocardiography, were done in a local hospital. A gradual accumulation of pericardial effusion and granular ventricular septal effusion were observed. Buccal mucosal biopsy did not confirm the tentative diagnosis of cardiac amyloidosis.

Physical examination on admission revealed severe dyspnea (New York Heart Association class III), sinus tachycardia (105 bpm), and hypotension (90/60 mm Hg). Laboratory tests showed high levels of N-terminal pro-B-type natriuretic peptide, characteristic of heart failure. Left ventricular wall thickening, left atrial enlargement, nonhomogeneous intraperitoneum, mitral restriction profile, and slightly reduced ejection fraction (45%) were observed on transthoracic echocardiography. The amount of pericardial fluid increased to 30 mm behind the posterior wall. On 24-hour Holter monitoring, multiple episodes of atrial tachycardia and a short single ventricular tachycardia were recorded. An estimated risk of sudden cardiac death (SCD) was 3.99%. Except for cardiovascular abnormalities, no other symptoms were observed.

Magnetic resonance imaging (CMR) was performed for further risk stratification. It showed heterogeneous edema with a perfusion disorder. The diagnostic workup was extended to include a myocardial biopsy. Histological staining of Congo Red (FIGURE 1A), immunohistological evaluation (FIGURE 1B), and transmission electron microscopy (FIGURE 1C) showed amyloid deposits.

The final diagnosis of cardiac amyloidosis was established, and the patient was transferred to a hematology department for further examination and treatment. After bone marrow biopsy

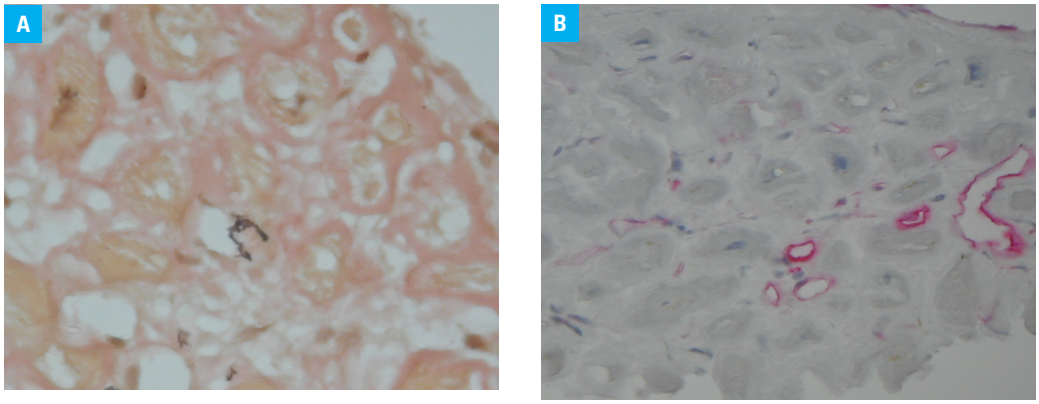

FIGURE 1 A – amyloid deposits: light pink amorphous material in the section of myocardial biopsy stained with Congo red; B – expression of human leukocyte antigens (HLA)-DR by the microvascular endothelium in immunostaining for HLA class II

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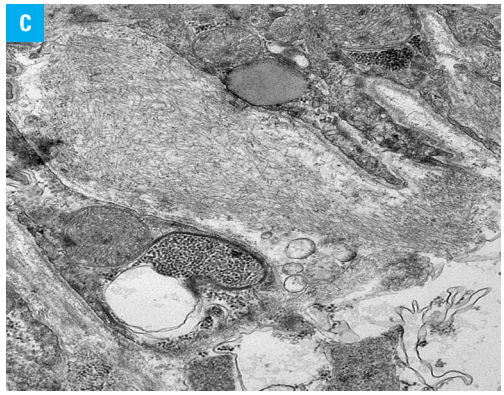

FIGURE 1
C – transmission electron micrograph showing randomly oriented straight fibrils adjacent to the sarcolemmal membrane

confirming light chain disease, chemotherapy with cyclophosphamide–bortezomib–dexamethasone followed by melphalan–dexamethasone regimen was applied. The therapy resulted in a gradual clinical improvement and a reduction in the amount of pericardial fluid.

A few months after chemotherapy, the patient was admitted to a cardiology department after an episode of syncope. Echocardiography revealed severe tricuspid valve regurgitation, moderate mitral insufficiency, impaired left ventricular function, and stable amount of pericardial fluid. Atrial and ventricular arrhythmias (single and couplets) were recorded on 24-hour Holter monitoring. Because exclusion of the arrhythmic origin of syncope episodes was not possible, the patient was referred for implantable cardioverter-defibrillator (ICD) implantation. Currently, she is in clinical remission. The implanted device recorded a few successfully treated episodes of ventricular fibrillation.

The present case shows that myocardial biopsy enables a diagnosis of cardiac amyloidosis and facilitates therapeutic decision making. Primary cardiac localization is often a diagnostic challenge.[1] A CMR image can by untypical. An invasive examination such as myocardial biopsy may be the only way to establish a proper diagnosis and choose adequate hematological treatment.[2] Furthermore, the presence of arrhythmia should prompt the treating physician to consider ICD implantation, even in patients with preserved ejection fraction, due to limitations of the SCD risk score.

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