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Echo solves clinical puzzle — cardiac amyloidosis case report

Echo rozwiązuje kliniczną zagadkę — opis przypadku amyloidozy serca

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Abstract

We present the case of a 53-year-old woman with New York Heart Association (NYHA) class III heart failure, despite optimal pharmacological therapy. Her medical history included: myocardial infarction with nonobstructive coronary arteries (MINOCA), heart failure with reduced ejection fraction and quadrantanopia with sensory neuropathy diagnosed as ischaemic stroke. Transthoracic echocardiography showed significant left ventricular hypertrophy with a 'granular' myocardial texture, global systolic dysfunction and restrictive mitral filling pattern. Two-dimensional (2D) transthoracic echo strain analysis demonstrated decreased global longitudinal strain with apical sparing, and helped to fit all the pieces of the jigsaw together to reach a consistent clinical diagnosis of cardiac amyloidosis.

Key words: 2D transthoracic echocardiography, cardiac amyloidosis, speckle tracking echocardiography

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Case report

A 53-year-old woman presented to the cardiology department with shortness of breath, orthopnoea, palpitations and New York Heart Association (NYHA) class III heart failure of a few months' duration, despite optimal pharmacological therapy with beta-blockers, angiotensin-converting enzyme inhibitors (ACEI), diuretics and digoxin. Three years earlier she had been hospitalised in our department due to exertional dyspnoea, slightly elevated troponins, and a reduced left ventricular ejection fraction (LVEF) of 30%. Coronary angiography performed at the time did not reveal coronary artery disease. She was diagnosed with myocardial infarction with nonobstructive coronary arteries (MINOCA) and dilated cardiomyopathy (DCM). One and a half years before that she was also hospitalised at a neurology department due to left superior quadrantanopia, dizziness, and numbness of the right upper extremity and right cheek. Computed tomography of the brain detected post-ischaemic injury, and she was treated conservatively for acute ischaemic stroke; all moderate symptoms persisted.

Physical examination on admission revealed regular pulse 65 bpm, normal blood pressure of 110/70 mm Hg, and moderate lower leg oedema. Her body mass index was 21 kg/cm^2. Six-minute walk test (6MWT) distance result was 150 metres and N-terminal pro-B-type natriuretic peptide (NT-proBNP) level was 7474 pg/mL, normal troponin T and creatine kinase myocardial bound (CK-MB). Transthoracic echocardiography showed significant left ventricular hypertrophy (LVH) [left ventricle (LVMI) mass index 160 g/m^2] with a 'granular' myocardial texture (Figure 1), global systolic dysfunction with left ventricular ejection fraction (LVEF) of 30%, restrictive mitral filling pattern (E/A 2.6, E/E' 20) with normal left atrial (LA) size [LA diameter 38 mm, left atrial volume index (LAVI) 32 mL/m^2]. Right ventricular function was low [tricuspid annular plane systolic excursion (TAPSE) 15 mm] with inferior vena cava diameter of 17 mm without respiratory variation. Twelve-lead electrocardiography (ECG) showed sinus rhythm with low voltage in limb leads

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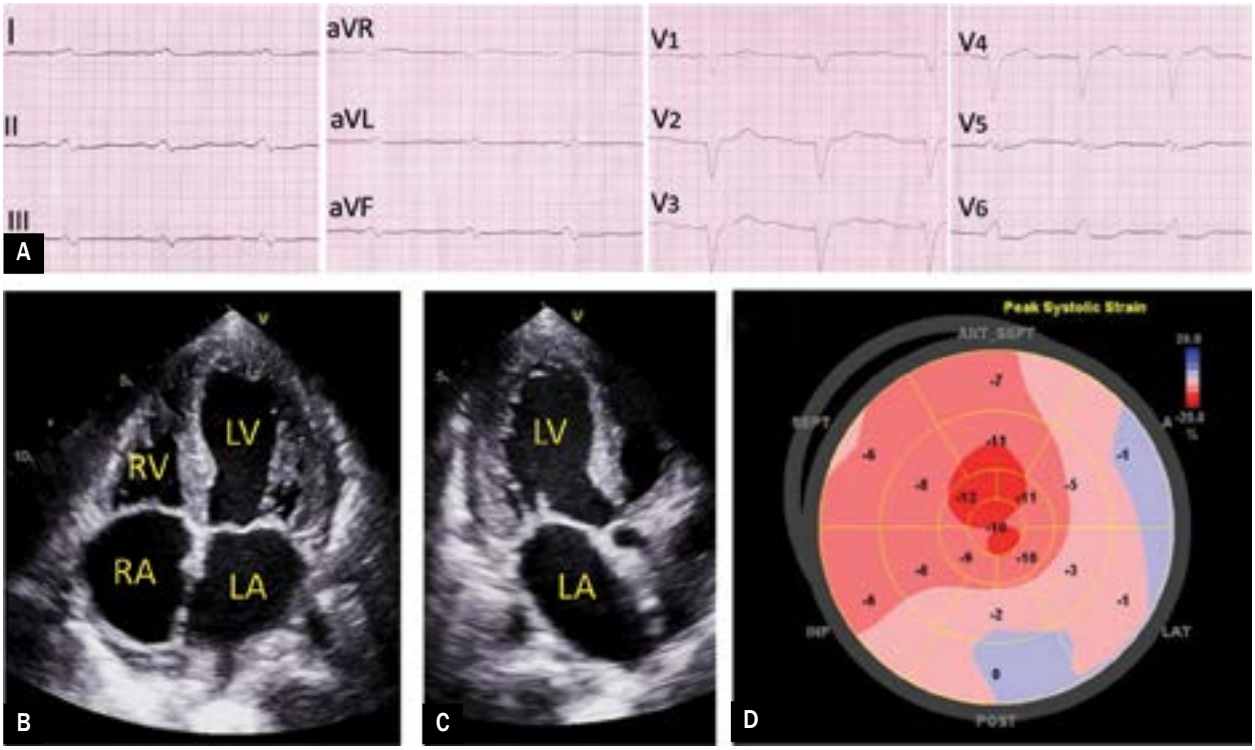

Figure 1A. Twelve-lead electrocardiography shows low voltage in limb leads; **B, C.** Apical four-chamber and three-chamber views (respectively) with increased left ventricular wall thickness and 'granular' texture of myocardium; **D.** Bull's-eye map of longitudinal strain with apical sparing; RV — right ventricle; LV — left ventricle; RA — right atrium; LA — left atrium

and no features of ventricular hypertrophy, incomplete left bundle branch block (LBBB).

This surprising discrepancy between transthoracic echocardiography (TTE) and ECG generated a suspicion of amyloidosis. Two-dimensional speckle tracking echocardiography demonstrated severely decreased global longitudinal strain –6.9% with less abnormal values of apical segments (apical sparing, the so-called 'cherry on top' pattern). Cardiac magnetic resonance imaging showed subendocardial circumferential late gadolinium enhancement, especially in basal segments of LV, as well as amyloid deposition in both atria. Serum protein immunofixation-electrophoresis detected a monoclonal band defined as immunoglobulin IgA-lambda.

Treatment was adjusted and a subcutaneous fat biopsy postponed at the patient's request. After rapid clinical deterioration, she died at home a few days later.

Discussion

Amyloidosis results from extracellular deposition of insoluble misfold fibrils. Cardiac involvement is observed in about 50–60% of patients with systemic primary amyloidosis (AL, amyloid light chain) [1] and is also common in other variants (ATTR, amyloid transthyretine). Differentiating subtypes involves a search for immune cell dyscrasia, myocardial scintigraphy with bone tracers, and cardiac biopsy. A diagnosis of AL type should prompt a haematology consult for specific treatment.

Our patient's puzzling medical history featured several significant elements. Firstly, the history of myocardial infarction with nonobstructive coronary arteries (MINOCA) — it is known that elevated troponin level may occur in amyloidosis [1]. Secondly — limb numbness, parasthesis and hypotension, recognised as ischaemic stroke, could represent misdiagnosed sensory and autonomic neuropathy, often occurring in patients with amyloidosis [2]; carpal tunnel syndrome and spinal canal stenosis are also common. Thirdly, heart failure can be a result of cardiac amyloidosis characterised by restrictive cardiomyopathy progressing to late-stage congestive heart failure with reduced ejection fraction [3]. Low voltage ECG with increased LV wall thickness, which is also typical for storage diseases, can be a whistleblower.

Classical features of amyloidosis in TTE include: symmetric LV and right ventricle (RV) thickening with normal to small cavity size, valvular thickening, granular myocardial texture, biatrial dilatation, pericardial effusion, progressive diastolic dysfunction and impairment of global longitudinal systolic (GLS) function with a relative 'apical sparing' pattern [3, 4].

Due to the unexpected clinical course, we cannot prove our diagnosis with a histology specimen, but this case presents a set of diagnostic features typical for cardiac amyloidosis. Importantly, strain analysis by TTE helped to fit the pieces of the jigsaw together to form a consistent clinical diagnosis. The recognition of amyloidosis is increasingly important as specific treatment options, such as tafamidis, start to emerge.

Conflict(s) of interest

The authors declare no conflict of interest.

Streszczenie

Przedstawiono opis przypadku 53-letniej kobiety z obniżoną tolerancją wysiłku fizycznego w III klasie czynnościowej według *New York Heart Association* (NYHA), mimo optymalnej farmakoterapii. W wywiadzie stwierdzono: przebyty zawał serca bez istotnych zmian w nasierdziowych tętnicach wieńcowych (MINOCA), niewydolność serca z obniżoną frakcją wyrzutową lewej komory oraz niedowidzenie kwadrantowe z neuropatią czuciową zdiagnozowane jako udar niedokrwienny mózgu. W echokardiograficznym badaniu przezklatkowym (TTE) uwidoczniono „ziarnistą" strukturę oraz znaczny przerost mięśnia sercowego lewej komory, uogólnioną dysfunkcję skurczową oraz restrykcyjny profil napływu mitralnego. Analiza dwuwymiarowa (2D) TTE wykazała zmniejszenie globalnego odkształcenia podłużnego z lepszą funkcją w obrębie koniuszka. Charakterystyczny obraz echokardiograficzny przyczynił się do złożenia wszystkich elementów klinicznych w diagnozę amyloidozy serca.

Słowa kluczowe: 2D echokardiografia przezklatkowa, amyloidoza serca, metoda śledzenia markerów akustycznych

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