Multimodality imaging-based diagnosis of cardiac transthyretin amyloidosis

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Introduction: Transthyretin amyloidosis cardiomyopathy (ATTR-CM) is a rare, progressive, life-threatening, hereditary disorder induced by a misfolded precursor protein, caused by mutations in the transthyretin gene. ATTR-CM is a challenging disease to recognize in early stages owing to its multisystem and nonspecific manifestations[1-3].

Case report: 65-year-old patient was hospitalized for the second time to our Clinic for Cardiovascular diseases in April 2022. Previous hospitalization happened 5 years ago and there was no ambulatory cardiology check-up between hospitalizations. He had progressive dyspnea on mild exertion and lower extremity edema. Therapy at home was furosemide occasionally. In past medical history he had left carpal tunnel surgery and Covid 19 infection in April 2022. On arrival blood pressure was 140/80 mmHg, pulse 100 beats per minute, absolutely arrhythmic. Basal weakened breath sounds, and leg edema were present. Troponin and NT pro BNP were elevated (91 ng/l and 3222 ng/l). 12-lead electrocardiogram showed peripheral micro-voltage, atrial fibrillation with ventricular rate around 110 bpm. Biatrial enlargement, increased left and right wall thickness, thickened papillary muscles, mildly reduced left ventricular ejection fraction and mono-phasic transmittal flow was found on transthoracic echocardiography. Testing for Fabry disease was negative. Cardiac magnetic resonance (CMR) found morphological changes and the pattern of contrast accumulation that suggested cardiac amyloidosis. Immunofixation electrophoresis showed no monoclonal (M) spike, gammopathy was unlikely. A biopsy of the buccal mucosa was performed, no amyloid deposits were found. Bone scintigraphy found accumulation of labelled hydroxydiphosphonate (HDP) that was visible in the myocardium, which points to ATTR-CM. Genetic testing is in progress.

Conclusion: ATTR-CM requires a high index of suspicion, and it should be suspected in patients with LV hypertrophy and heart failure. The diagnosis of cardiac amyloidosis requires a combination of multi-modality imaging including echocardiography, CMR and scintigraphy. An imaging modality that can accurately diagnose ATTR-CM without the need for invasive cardiac biopsy is nuclear scintigraphy using bone-avid radio-tracers[4]. Timely diagnosis is important since the treatment is possible and improves prognosis in these patients.

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