Transthyretin amyloid cardiomyopathy – challenges in diagnostic and therapeutic protocol

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Goal: To present protocol for diagnosis and differentiation of cardiac amyloidosis, and to review treatment options for patients with transthyretin amyloid cardiomyopathy (ATTR-CM) in daily clinical practice.

Case presentation: 70-year-old man was hospitalized with symptoms consistent with peripheral neuropathy, polyarthralgia, Raynaud syndrome and dyspnea on exertion. His past medical history included hypertension and hypothyroidism. Electrocardiogram (ECG) revealed left axis deviation, low voltage and intermittent atrial fibrillation. Echocardiography showed concentric left ventricular hypertrophy with granular speckling, normal ejection fraction and grade 1 diastolic dysfunction. Moderate aortic stenosis with mild aortic regurgitation was present, as well as moderate mitral regurgitation and mild tricuspid regurgitation. Subsequent analysis of global longitudinal strain (GLS) showed GLS reduction with nonspecific apical sparing pattern (**Figure 1**). Infiltrative cardiomyopathy was suspected, and we obtained serum and urine protein electrophoresis with immunofixation and the kappa/lambda ratio, which were negative for clonal plasma cell dyscrasia. Bone scintigraphy showed Technetium 99m-methyl diphosphonate uptake in the myocardium which was less/similar to ribs uptake (Perugini grade 1-2). Cardiac magnetic resonance (CMR) was obtained since scintigraphy results were inconclusive, and it confirmed cardiac amyloidosis pattern. Patient had significantly high levels of rheumatoid factor, without specific diagnostic criteria for rheumatological diseases. Electromyoneurography of arms and legs confirmed severe polyneuropathy. Since the patient presented with both cardiomyopathy and neuropathy, disease-specific therapies to consider would be tafamidis and patisiran. Genetic testing should be performed to detect specific mutations and to guide the treatment.

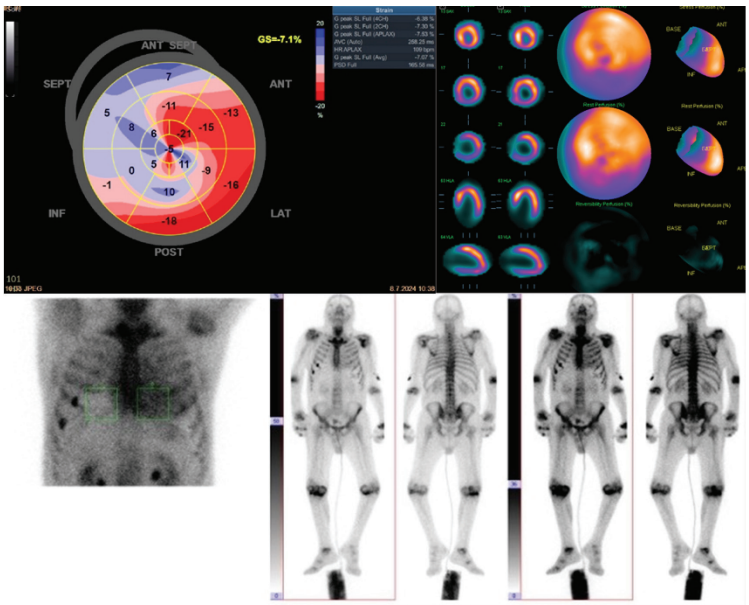

FIGURE 1. Analysis of global longitudinal strain (GLS) showed GLS reduction (-7.1%) without typical pattern. Perugini grade 2 with a region of interest (ROI) ratio of 1.51.

Conclusion: Extracardiac manifestations which represent 'red flags' for ATTR-CM, should guide clinicians to perform echocardiography. After establishing the diagnosis, patient should be treated with specific treatment based on the dominant disease phenotype.[1-3]

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LITERATURE

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