Cardiac Amyloidosis with Mild Left Ventricular Hypertrophy

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Introduction:
Amyloidosis describes a disease caused by pathologic deposition of misfolded proteins. It is a multi-organ disease presenting with unspecific symptoms and diagnosis is often delayed.

Case presentation:
A 66-year-old male patient was admitted to the ER complaining of progressive exertional dyspnea grade III for 6 weeks, orthopnea for 3 weeks and bilateral lower limb edema for 1 month. Past medical history: right pleural effusion with pleurocentesis 1m before. ECG showed low QRS voltage relatively disproportionate to LV thickness with a pseudo infarction pattern. His Echo showed Unexplained biventricular hypertrophy with thickening of A-V valves and Intra atrial septum, thin rim of pericardial effusion and grade II diastolic dysfunction with elevated filling pressures. Speckle tracking showed reduced global longitudinal strain with relative "apical sparing". His cardiac MRI showed difficulty in differentiation between the myocardium and the blood pool denoting failure of nulling, diffuse non-territorial patchy mid myocardial and sub myocardial enhancement. Native T1 mapping values were elevated in all myocardial segments with increased ECV > 50 % in most of the LV segments. His serum protein electrophoresis had shown high amyloid A protein. His serum light chain assay showed an elevated free lambda chain with a kappa/lambda ratio of 0.013. Abdominal fat biopsy showed positive amyloid deposits AL (lambda) and negative amyloid deposits (KAPPA). In the fat biopsy, the caongo red cell was negative. The patient was diagnosed with AL amyloidosis associated with multiple myeloma.

Discussion:
Amyloidosis results from the deposition of misfolded proteins affecting multiple organs. Types of amyloidosis affecting the heart include AL amyloidosis which is caused by deposition of light chain immunoglobulin and represents 10 % with multiple myeloma, TTR amyloidosis caused by a gene mutation in the TTR gene or age-related process and AA amyloidosis which is a genetic predisposition, that mainly affects GIT and kidney (rarely affects the heart). Diagnosis of amyloidosis needs high clinical suspicion. Red flags to suspect amyloidosis include heart failure symptoms, orthostatic hypotension, carpal tunnel syndrome, low voltage QRS in ECG, unexplained LVH and diffuse late gadolinium enhancement. Screening for amyloidosis is done by serum or blood immunofixation electrophoresis and kappa/ lambda free light chain ratio. Diagnosis is confirmed by tissue biopsy from abdominal fat and affected organ with congo red staining.

Conclusion:

- Amyloidosis is a multi-organ disease presenting with unspecific symptoms of different severity.
- Diagnosis is delayed and needs high clinical suspicion.
- Diagnosis needs a multimodality approach including clinical, imaging (Echo – CMR) and histopathology.
- Diagnosis and treatment need a multidisciplinary team with cooperation to improve the quality of management.

Keywords:
Amyloidosis, multimodality diagnosis

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