It's Just Diastolic Heart Failure, Right? A Lesson in Humility from a Patient with TTR-Amyloid Cardiomyopathy.

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It's Just Diastolic Heart Failure, Right? A Lesson in Humility from a Patient with TTR-Amyloid Cardiomyopathy

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INTRODUCTION

Diastolic heart failure (dCHF) is a frequent diagnosis which can have many etiologies. The diagnosis is often attributed to hypertensive heart disease and further workup of the cause is disregarded. Systemic senile transthyretin amyloidosis, with a prevalence of 6-11 cases per million person-years, can often deposit in cardiac tissue. Cardiac amyloidosis (CA) associated with senile transthyretin deposition has been found in up to 13% of those with dCHF. However, many cases of dCHF caused by transthyretin related (TTR) CA may go undiagnosed.

CASE

A 64-year-old male presented to our ED with worsening dyspnea on exertion and weight gain. His past medical history included NYHA stage 3, diastolic heart failure without coronary artery disease or a family history of heart failure, paroxysmal orthostatic tachycardia syndrome (POTS), carpal tunnel, asthma, and multiple antihypertensive medication intolerances. He had already had five admissions over the last five years for exacerbations of dCHF.

A repeat ECHO demonstrated severe concentric left ventricular hypertrophy with global hypokinesis, an EF of 45% and elevated diastolic filling pressures. Repeat ECG was notable for lower than expected voltage. Given his progressively worsening concentric LVH, lower than expected voltage on ECG and progressively worsening LV function, infiltrative disease needed to be ruled out. The patient underwent cardiac MRI (image 1) which confirmed severe concentric LV hypertrophy with late gadolinium enhancement within the LV myocardium suggesting infiltrative cardiomyopathy. He was scheduled for a myocardial biopsy with congo red staining, and the diagnosis of cardiac amyloidosis (CA) was confirmed (image 2). Due to the patient's negative urinary and serum protein electrophoresis, a 99m Tc-pyrophosphate scintigraphy was performed to assess for TTR CA. The scan confirmed the diagnosis with notable uptake within the LV myocardium consistent with TTR CA (image 3). Eventually, pathologic evaluation confirmed the diagnosis of TTR CA, and amino acid testing was negative for known TTR mutations suggesting the senile form of the TTR CA. The patient is currently being evaluated for heart transplantation.

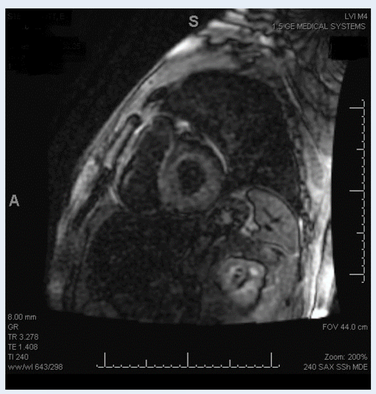

Image 1: Cardiac MR showing severe concentric left ventricular hypertrophy with late gadolinium enhancement.

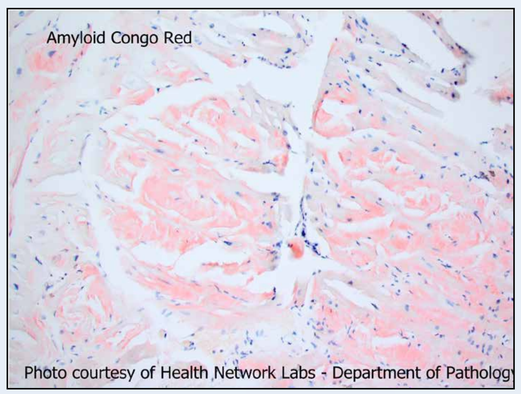

Image 2: Congo Red stain of the amyloid deposition in the myocardium.

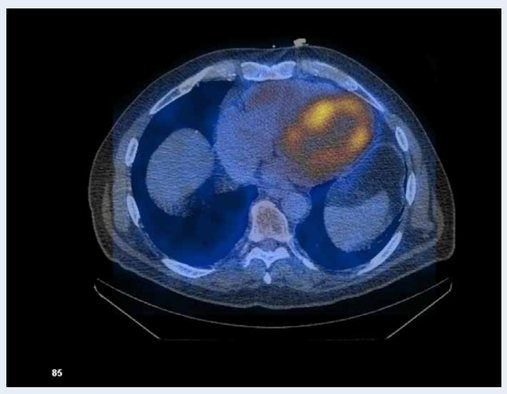

Image 3: 99m Tc-pyrophosphate scintigraphy with notable uptake in the myocardium of the left ventricle suggestive of TTR amyloidosis.

DISCUSSION

- Even though dCHF is a common cause of hospitalization, the initial differential for the cause of the heart failure must remain broad. The diagnosis of TTR-CA in patients with dCHF should be considered to avoid delay in optimizing treatment.
- Patients with left ventricular concentric hypertrophy could be due to TTR-CA and could benefit from a more aggressive workup.
- This patient was able to benefit by starting the transplant evaluation process as well as avoiding typical heart failure medications which could potentially aggravate his condition.
- As clinicians it is easy to be caught up in diagnosing and treating what is common and known. It is important to consider all possible etiologies to assist in coming to the correct diagnosis in a timely manner.

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