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Case Report

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Cardiac Amyloidosis: The Forgotten Disease

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Cardiac amyloidosis leads to signs and symptoms of heart failure. A clinical condition when one of more than 30 different precursor proteins with unstable tertiary structure, misfolds and aggregates as insoluble amyloid fibrils which deposit in the extracellular space of organs and soft tissue. Among the many types of amyloidosis, nearly all cases of clinical cardiac amyloidosis (95 percent) are due to transthyretin amyloidosis (ATTR) or light chain amyloidosis. Studies suggest that wild-type transthyretin amyloidosis (ATTRw) is an underdiagnosed cause of heart failure, with a prevalence of 13 percent in a cohort of patients presenting with clinical manifestations of HF with preserved ejection fraction [1,2].

This is the case of a 77 years-old male with a past medical history of arterial hypertension, atrial- fibrillation, dyslipidemia, and peripheral vascular disease. The patient visited a previous institution with the chief complaint of dyspnea on minimal exertion and bilateral pitting edema that has been progressive since a week ago. The patient was transferred to our institution where the physical examination was remarkable for distant heart sounds, bilateral basilar crackles during auscultation, and bilateral pitting edema without JVD. Chest X-Ray showed bilateral effusion and boot-shaped heart that goes with cardiac effusion. ECG is remarkable for atrial fibrillation with an adequate ventricular response, left ventricular hypertrophy, and low voltage [3,4].

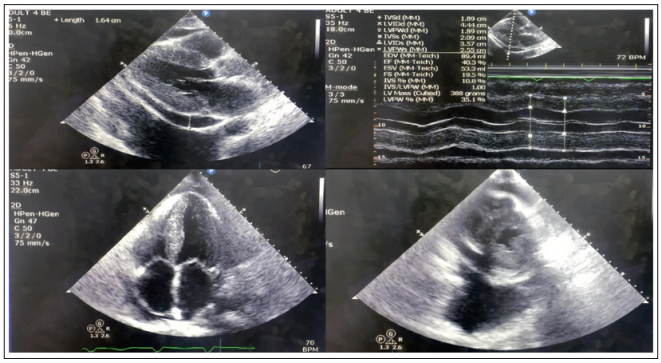

Figure 1: Echocardiogram resulted in preserved heart failure, the existence of pericardial fluid, biventricular and biatrial enlargement. The suspected hypertrophy was confirmed by the left ventricular mass index (based on BMI, gender, and age) in his case more than 115 g/m2 (226 g/m2) and the relative wall thickness of more than 0.42 that goes with concentric hypertrophy. Studies report that predominant hallmarks of cardiac amyloidosis are discordance between increased left ventricular wall thickness and QRS low- voltage; it is crucial to highlight that our patient did not present the constant characteristic of amyloidosis in echocardiogram as the relative apical sparing of longitudinal strain or the existence of sparkling myocardium. The patient underwent right heart catheterization to assess the possibility of pulmonary hypertension as the cause of the concentric hypertrophy but resulted within normal hemodynamics parameters. Laboratories to rule out light-chain amyloidosis and hemochromatosis as the cause of the concentric hypertrophy resulted with M spike not observable, free urine and serum Kappa and lambda chains undetectable, iron level normal, and ferritin normal levels.

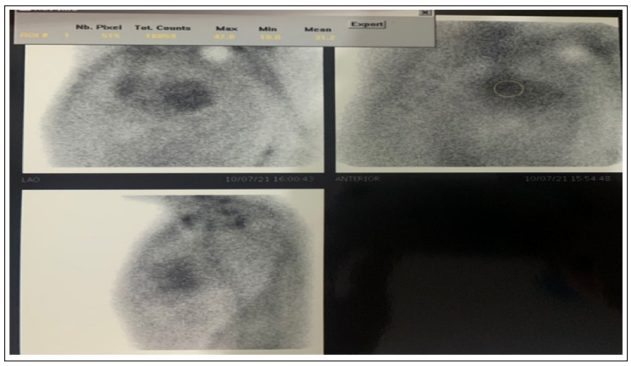

Figure 2: The diagnostic utility of 99mTechnetium-Pyrophosphate (PYP) Imaging for Transthyretin Cardiac Amyloidosis was positive. Perugini staging system based on simple visual scoring of the three-hour planar image: grade 0 being negative (no cardiac uptake) and grades 1 to 3 defined as detection of progressively greater cardiac uptake and decrease in the bone uptake, based on more than 1.5 heart/contralateral lung ratio.

Diagnosis without the requirement of an invasive diagnostic test was done by the 99mTechnetium-Pyrophosphate (PYP) Imaging for Transthyretin Cardiac Amyloidosis in the absence of serum and urine light chains. Unfortunately, the patient died before treatment. Finally, physicians should be aware that cardiac amyloidosis is a life- threatening, progressive disease often underdiagnosed and misdiagnosed. Untreated cases have a median survival of approximately 2 to 3.5 years, accurate, early diagnosis of non-invasive methods is the key to enabling appropriate patient care.

Conflicts of Interest
None of the authors have any relevant financial disclosures

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