When Two Paths Cross: A Case of Transthyretin Cardiac Amyloidosis in a Patient with Multiple Myeloma

Dhruv Patel DO[1], Rahul Patel DO[2], Arjun Bagai DO[1], Andrew Mariano DO[2], Tarun Jain MD[2], Seban Liu DO[2], Vijay M Haryani MD[2], Abimanyu Beri MD[2], Nha Huynh DO[2]

[1] Riverside Medical Center, Department of Internal Medicine, Kankakee, IL ; [2] Riverside Medical Center, Heart and Vascular Institute, Kankakee, IL

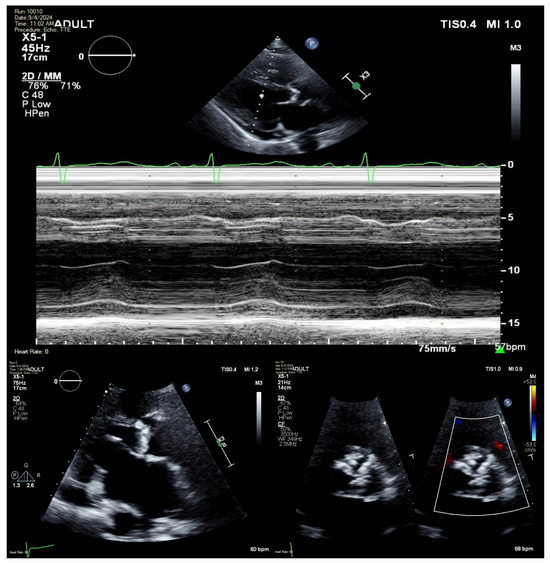
BACKGROUND

Cardiac amyloidosis (CA) is caused by deposition of misfolded proteins and is broadly classified into two source categories: AL amyloidosis (amyloid light chain from plasma cell dyscrasia) and TTR amyloidosis (misfolding of transthyretin). We report a rare case of a patient with active multiple myeloma with concomitant TTR CA.

CASE PRESENTATION

A 77-year-old male with a history of systolic heart failure, persistent atrial fibrillation, CAD, IgA kappa multiple myeloma was admitted for heart failure exacerbation and atrial fibrillation with rapid ventricular rate. Initial testing showed kappa to lambda FLC ratio of 16.07. Echocardiogram was obtained showing LVEF of 10% with low-flow, low-gradient aortic stenosis. Rate and rhythm control therapy was initiated, and the patient was stabilized and underwent left heart catheterization which revealed severe multivessel disease. The patient agreed to undergoing a CABG procedure with surgical aortic valve replacement. A repeat echocardiogram was performed which showed improvement in LVEF to 40-45% and presence of severely reduced longitudinal strain with an apical sparing pattern raising concern for possible cardiac amyloidosis. The patient subsequently underwent a CABG procedure and surgical aortic valve replacement. Biopsy of myocardium and left atrial appendage were sent to pathology during the procedure. Histopathological report of the cardiac tissue sample confirmed the presence of amyloid deposits. Mass spectrometry confirmed TTR amyloidosis. The patient was subsequently discharged with a plan to initiate tafamidis as an outpatient.

FIGURE 1



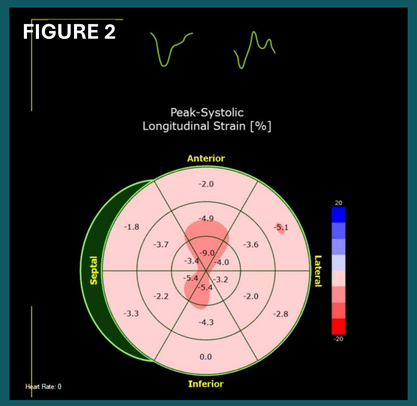
M-mode echocardiography of parasternal long-axis view demonstrates mild to moderately reduced left ventricular systolic function, thickened valves, and small pericardial effusion. Severely calcified trileaflet aortic valve further seen in both parasternal long-axis and short-axis view

DISCLOSURE INFORMATION

The authors have no disclosures.

Cardiac amyloidosis is **an underdiagnosed condition** that necessitates a **heightened clinical suspicion** for proper identification.

Our case highlights the importance of **proper diagnostic testing** in elderly patients with history of light chain disease to ensure an **accurate diagnosis of cardiac amyloidosis** subtype



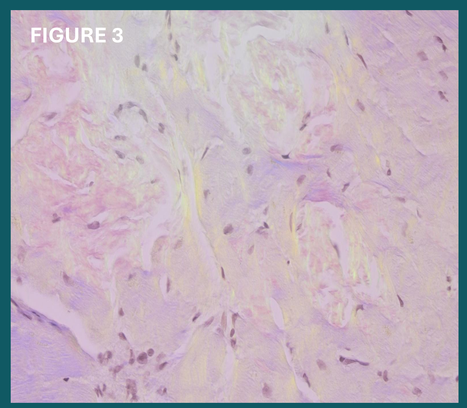
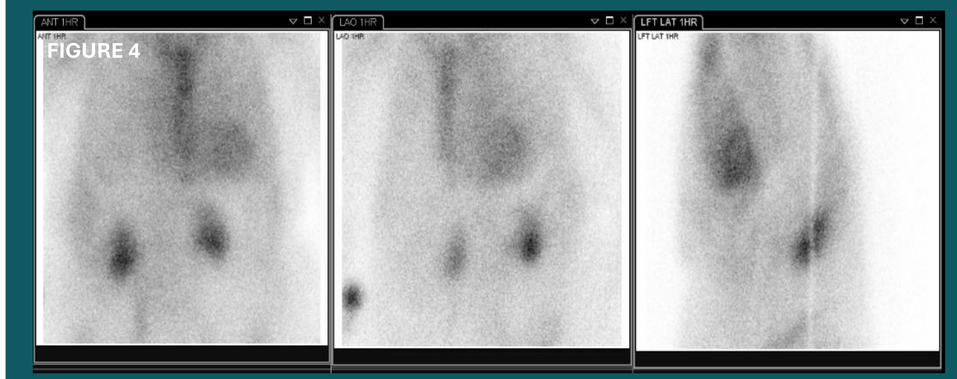


Figure 2: Severely reduced global longitudinal strain with apical sparing pattern suspicious for cardiac amyloidosis

Figure 3: Biopsy of the left atrial appendage demonstrating apple-green birefringence on congo red stain consistent with cardiac amyloidosis

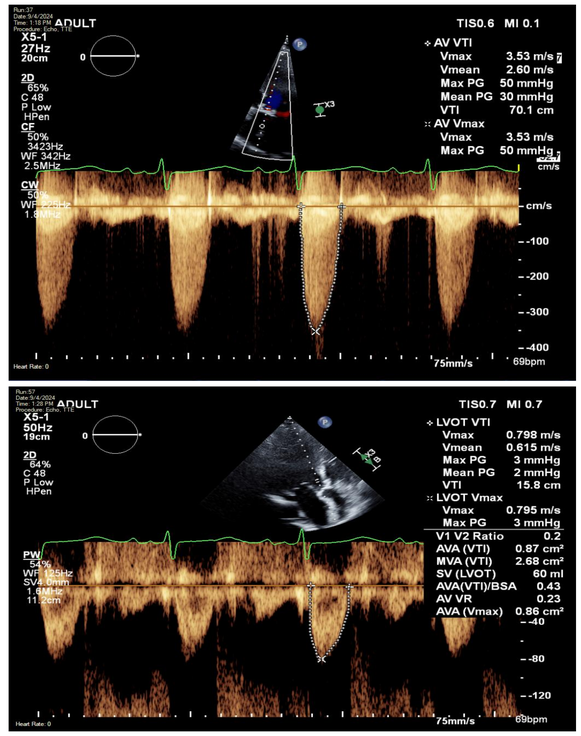


1-hour ^{99m}Tc-PYP planar and SPECT amyloid scan demonstrates diffuse tracer uptake in the myocardium with a quantitative ratio of myocardial activity to background chest activity greater than 1.5, visualized subjective ratio greater than 2, a semiquantitative visual score of grade 3

DISCUSSION

The reported prevalence of CA in the United States is approximately 55 cases per 100,000 individuals Studies have shown that the prevalence may be as high as 20-25% in elderly patients with heart failure and left ventricular thickening with most cases consisting of ATTR amyloidosis (85%) and a minority consisting of AL amyloidosis (2%). A PYP scan demonstrates an 86% specificity for CA attributed to ATTR, but false-positive results may occur in patients with AL amyloidosis. For this reason, serum and urine immunofixation are performed prior to obtaining imaging. Other imaging modalities such as F-labelled PET scans show promising findings for differentiating AL versus ATTR cardiac amyloidosis, however no large size studies have been performed to confirm their efficacy

FIGURE 5



Continuous wave doppler of aortic valve and pulse wave doppler of left ventricular outflow tract demonstrating severe low-flow, low-gradient aortic valve stenosis (Vmax 3.53 m/s, mean gradient 30 mmHg, DI 0.23, AVA 0.87 cm^2, SVi 30 ml/m^2, LVOT 2.2 cm)

REFERENCES

Yamamoto, Y., Onoguchi, M et al. (2012). Novel method for quantitative evaluation of cardiac amyloidosis using (201)TlCl and (99m)Tc-PYP SPECT. *Annals of nuclear medicine*, *26*(8), 634–643.

Bokhari, S et al. (2013). (99m)Tc-pyrophosphate scintigraphy for differentiating light-chain cardiac amyloidosis from the transthyretin-related familial and senile cardiac amyloidoses. *Circulation. Cardiovascular imaging*, *6*(2), 195–201.

Baker K. R. (2022). Light Chain Amyloidosis: Epidemiology, Staging, and Prognostication. *Methodist DeBakey cardiovascular journal*, *18*(2), 27–35.