

Red Flags of Transthyretin Amyloid Cardiomyopathy  
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Abstract

Background

Cardiac amyloidosis is historically defined as a rare disease; however, it is commonly misdiagnosed. There are two common types of cardiac amyloidosis: light-chain (AL) and transthyretin amyloid cardiomyopathy (ATTR-CM). AL amyloidosis is caused by overproduction of immunoglobulin light-chain fragments and is treated with chemotherapy and/or targeted therapy. It is a fast-paced disease and is classified as an emergency. ATTR-CM can occur with aging or a hereditary gene mutation. It can present as heart failure with preserved ejection fraction (HFpEF), aortic stenosis, cardiac arrhythmias, bilateral carpal tunnel syndrome, lumbar spinal stenosis, biceps tendon rupture, and increased LV wall thickness. These symptoms are considered red flags for ATTR-CM. Often, cardiac amyloidosis goes undiagnosed because it can easily be misdiagnosed as one of the red flag symptoms. ATTR amyloidosis is treated with tafamidis, a medication indicated in adults with ATTR-CM to reduce cardiovascular mortality and cardiovascular-related hospitalization.

Methods

This study looked for the presence of ATTR-CM red flag symptoms in patients who underwent transcatheter aortic valve replacement (TAVR) including HFpEF, bilateral carpal tunnel syndrome, lumbar spinal stenosis, biceps tendon rupture, and increased LV wall thickness. It was also documented if patients had a diagnosis of ATTR-CM or were prescribed tafamidis.

Results

There were 131 patients who were included in this study. Several red flag symptoms were identified; however, no patient had a diagnosis of ATTR-CM and there was no history of any patients taking tafamidis. HFpEF was the most commonly identified red flag symptom, found in 53 (40.5%) patients. Of the 133 red flag symptoms identified, 82 patients had at least one red flag symptom with 37 of those patients having more than one symptom.

Discussion

62% of patients had a history of at least one red flag symptom of ATTR-CM and 28% of patients experienced more than one red flag symptom. Over half of the population has experienced a red flag symptom, yet there were no ATTR-CM diagnoses identified. This could be due to the small sample size and the location of the study. Furthermore, ATTR-CM frequently presents as heart failure and 40.5% of the study population had a diagnosis of HFpEF, leading one to believe ATTR-CM could have been diagnosed as heart failure and not further worked up. Overall, this study did not provide results as hoped, but it

will help to bring awareness to the disease state and its presenting symptoms.

### Conclusion

The results of this study did not find any presence of ATTR-CM in patients who underwent a TAVR between January 2021 to December 2022. In the future, studies should be conducted at larger health systems that have a higher prevalence of ATTR-CM.