A multidisciplinary approach in the search for transthyretin amyloid cardiomyopathy

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Introduction: Transthyretin amyloid cardiomyopathy (ATTR-CM) is a rare but rapidly progressive disease. It is caused by the misfolding of transthyretin protein in the liver, resulting in the formation of amyloid fibrils. These harmful fibers deposit in the tissues of the carpal tunnel and may be one of the causes of carpal tunnel syndrome. While carpal tunnel syndrome can be an early symptom of ATTR amyloidosis and is a valuable indicator for screening for cardiac amyloidosis, it may not be sufficient. To improve the identification of patients at risk for ATTR-CM, we recommend the use of phenotypes obtained by a machine learning-adapted algorithm to identify possible transthyretin amyloid cardiomyopathy. The inclusion of cardiomegaly, osteoarthritis, and cardiovascular symptoms (such as heart failure, atrial fibrillation, and heart block) as part of the phenotypic criteria is a comprehensive approach.[1-3] We will present our first confirmed patient with ATTR-CM who meets all the above criteria.

Case report: 80-year-old patient was hospitalized for replacement of a pacemaker battery originally implanted in 2017 for bradycardic atrial fibrillation. The patient has several comorbidities, including hypertension, prostatic hyperplasia, and carpal tunnel surgery five years ago that resulted in residual polyneuropathy. In addition, the patient has an elevated NT-proBNP level (>1300pg/l) and the following echocardiographic findings: concentric biventricular hypertrophy, septal wall thickness >14 mm, biatrial enlargement, moderately severe mitral and tricuspid regurgitation, and a mildly reduced ejection fraction (EF 45%), corresponding to heart failure with mildly reduced ejection fraction. ATTR-CM was suspected and eventually confirmed by scintigraphy. The patient is scheduled for treatment with tafamidis.

Conclusion: Implementation of a screening program for ATTR-CM in patients who have previously undergone carpal tunnel release surgery and present with cardiovascular symptoms such as heart failure, atrial fibrillation, and heart block is a proactive and potentially life-saving approach to early detection and intervention. Collaboration with experts, ethical considerations, and validation of the screening algorithm are essential components of its success.

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LITERATURE

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