Severe aortic stenosis in a patient with transthyretin cardiac amyloidosis

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Introduction: Transthyretin cardiac amyloidosis (ATTR-CM) frequently coexists with aortic stenosis (AS), particularly in patients over 65 years, with a reported prevalence of 4–16%, especially among those undergoing transcatheter aortic valve implantation (TAVI). Identifying concomitant ATTR-CM in AS is challenging due to overlapping features, yet it is crucial for accurate diagnosis and optimal management[1].

Case report: 83-year-old male patient with a history of arterial hypertension, hyperlipidemia, and a previous cerebrovascular stroke was hospitalized due to progressive dyspnea and chest discomfort. Physical examination revealed a midsystolic precordial murmur and the presence of peripheral edema. Laboratory analysis demonstrated elevated levels of N-terminal pro–B-type natriuretic peptide. Transthoracic echocardiography showed a normal-sized left ventricle with concentrically thickened and hyperechoic walls (17 mm), preserved ejection fraction, but reduced global longitudinal strain with an apical sparing pattern and miminal pericardial effusion. Furthermore, there was an estimation of low-flow, low-gradient severe AS (AVA VTI 0.9 cm^2) with CT calculated calcium score of 1475. Coronary angiography showed no signs of coronary artery disease. Due to echocardiographic suspicion of cardiac amyloidosis, the patient underwent targeted diagnostic evaluation. Hematological workup excluded systemic light-chain (AL) amyloidosis. Tc-99m-pyrophosphate bone scintigraphy combined with SPECT/CT revealed intense radiotracer uptake in the myocardium, corresponding to a Perugini grade 3. This confirmed ATTR-CM diagnosis in our patient, which also explains relatively lower aortic valve calcium score than expected. After Heart Team discussion, the patient was accepted for TAVI procedure, which was later performed successfully. The patient remained stable during six months follow-up.

Conclusion: Patients with concomitant AS and ATTR-CM exhibit significantly poorer clinical outcomes compared to those with isolated AS[2], underscoring the importance of early recognition and tailored management of this high-risk subgroup. Echocardiography has a crucial role in identifying early signs of ATTR-CM, which should prompt further diagnostic evaluation.

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LITERATURE

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