Abstract

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FADOI The serial accumulator

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Background

A 74 year old lady came to our with non-productive cough, sloping edema and worsening dyspnoea for about 10 days, she had already performed a chest CT scan at home which showed slight bilateral baseline fluid. She decided to go to the ED where they found a mild normocytic normochromic anemia (Hb 11.4 g/dL), Troponin I 148 ng/L, BNP 2554 pg/mL, and LDH 330 U/L. The troponin curve performed with dosage at three and ten hours was not significant due to ischemic in progress. At the anamnestic interview, evidence of arterial hypertension, chronic ischemic heart disease, recent PMK implant (about 20 days from the onset of symptoms for BAV II° Mobitz 2), previous HCV infection subjected to eradicating antiviral therapy. On admission, the patient was dyspneic but hemodynamically stable. In the light of the suggestive picture of congestive heart failure, intravenous diuretic therapy was set up with furosemide and potassium canrenoate with consequent benefit on respiratory dynamics. On the fifth day of hospitalization, in order to monitor cardiac function, it was decided to perform a new echocardiogram, which showed severe concentric hypertrophy of the left ventricle with preserved volume, moderate mitral regurgitation and sclerocalcified aortic valve, concluding for probable storage disease. In view of the suspicion of cardiac infiltrative pathology, it was decided to perform an endomyocardial biopsy; the latter confirmed the presence of a picture compatible with storage disease, specifically attributable to "cardiac amyloidosis with myocytolysis". Immuno-electromicroscopy evidence of ultrastructural picture compatible with AL amyloidosis (lambda light chains).

Discussion

Cardiac amyloidosis is a severe and progressive infiltrative disease caused by hereditary or acquired physiological abnormalities that lead to the accumulation of amyloid fibrils in the heart. More than 98% of cardiac amyloidosis is caused by just two types of proteins: immunoglobulin light chains and transthyretin. Typically, cardiac amyloidosis manifests itself with a large number of extracardiac signs and symptoms, defined as "red flags", which can guide the diagnostic suspicion. These include proteinuria, macroglossia, bruising of the skin, and a history of carpal tunnel syndrome. But there are also cardiac red flags, such as the presence of heart failure with a level of NT-proBNP disproportionate to the echocardiographic evidence, right heart failure in the presence of normal valvular and ventricular function, persistently elevated troponin levels, a QRS voltage too low or an early conduction disturbance.

However, even in the absence of indicative signs and symptoms such as "red flags", a storage disease must always be suspected in patients with multiple ischemic and coronary events unscathed, and in acute decompensated patients. The therapeutic approaches are distinguished in those aimed at treating the comorbidities deriving from the disease, such as heart failure, arrhythmias, conduction disturbances, thromboembolism, aortic stenosis, and actual disease-modifying treatments which are able to act on different stages of production of amyloid fibrils, from formation to assembly, such as Tafamidis, a small molecule that acts by stabilizing the transthyretin tetramer.

Bibliografia

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