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Experience of Two Major Research Centers in the Diagnosis of Cardiac Amyloidosis from Transthyretin

Authors : Ioannis Panagiotopoulos, Aristidis Anastasakis, Konstantinos Toutouzas, Ioannis Iakovou, Charalampos Vlachopoulos, Vasilis Voudris, Georgios Tziomalos, Konstantinos Tsioufis, Efstathios Kastritis, Alexandros Briassoulis, Kimon Stamatelopoulos, Alexios Antonopoulos, Paraskevi Exadaktylou, Evanthia Giannoula, Anastasia Katinioti, Maria Kalantzi, Evangelos Leontiadis, Eftychia Smparouni, Ioannis Malakos, Nikolaos Aravanis, Argyrios Doumas, Maria Koutelou

Abstract : Introduction: Cardiac amyloidosis from Transthyretin (ATTR-CA) is an infiltrative disease characterized by the deposition of pathological transthyretin complexes in the myocardium. This study describes the characteristics of patients diagnosed with ATTR-CA from 2019 until present at the Nuclear Medicine Department of Onassis Cardiac Surgery Center and AHEPA Hospital. These centers have extensive experience in amyloidosis and modern technological equipment for its diagnosis. Materials and Methods: Records of consecutive patients (N=73) diagnosed with any type of amyloidosis were collected, analyzed, and prospectively followed. The diagnosis of amyloidosis was made using specific myocardial scintigraphy with Tc-99m DPD. Demographic characteristics, including age, gender, marital status, height, and weight, were collected in a database. Clinical characteristics, such as amyloidosis type (ATTR and AL), serum biomarkers (BNP, troponin), electrocardiographic findings, ultrasound findings, NYHA class, aortic valve replacement, device implants, and medication history, were also collected. Some of the most significant results are presented. Results: A total of 73 cases (86% male) were diagnosed with amyloidosis over four years. The mean age at diagnosis was 82 years, and the main symptom was dyspnea. Most patients suffered from ATTR-CA (65 vs. 8 with AL). Out of all the ATTR-CA patients, 61 were diagnosed with wild-type and 2 with two rare mutations. Twenty-eight patients had systemic amyloidosis with extracardiac involvement, and 32 patients had a history of bilateral carpal tunnel syndrome. Four patients had already developed polyneuropathy, and the diagnosis was confirmed by DPD scintigraphy, which is known for its high sensitivity. Among patients with isolated cardiac involvement, only 6 had left ventricular ejection fraction below 40%. The majority of ATTR patients underwent tafamidis treatment immediately after diagnosis. Conclusion: In conclusion, the experiences shared by the two centers and the continuous exchange of information provide valuable insights into the diagnosis and management of cardiac amyloidosis. Clinical suspicion of amyloidosis and early diagnostic approach are crucial, given the availability of non-invasive techniques. Cardiac scintigraphy with DPD can confirm the presence of the disease without the need for a biopsy. The ultimate goal still remains continuous education and awareness of clinical cardiologists so that this systemic and treatable disease can be diagnosed and certified promptly and treatment can begin as soon as possible.

Keywords : amyloidosis, diagnosis, myocardial scintigraphy, Tc-99m DPD, transthyretin
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