13. TRANSTHYRETIN CARDIAC AMYLOIDOSIS AND BONE SCINTIGRAPHY: STATE OF THE ART DETECTION IN RARE DISEASES

Author: Asavei Letiția; **Co-author:** Cârciumaru Marius; Ciocoiu-Bunilă Ioan Vlad; Gorea Diana Valentina

Scientific advisor: Ionescu Teodor Marian; Nuclear Medicine Specialist; University Assistant in Pathophysiology; *Gr. T. Popa* University of Medicine and Pharmacy, Iasi, Romania

Introduction. Transthyretin cardiac amyloidosis (ATTR-ca) is a rare protein deposition disease characterized by the accumulation of amyloid in the myocardium altering its function. Diagnosing it has proven to be a challenge even for the most experienced physician mainly because the symptoms that these patients present are common in more than one disease. Until this day, the gold standard method for diagnosing ATTR is endomyocardial biopsy. However, due to the risk that it presents and the fact that it is widely unavailable, a correlation between clinical and paraclinical investigations is recommended in such cases.

Case statement. We present the case of an 82 years old female patient, suspect of cardiac amyloidosis based upon electrocardiography and ultrasonography criteria. The patient was sent to the nuclear medicine laboratory of "Sf. Spiridon" County Emergency Hospital and underwent bone scintigraphy with a Siemens Dual-Head Gamma Camera equipped with a low energy, high resolution collimator. We acquired whole body images (early at 10 minutes and delayed at 2 hours) followed by static and SPECT centered on the thorax, 2 hours after the i.v. administration of 99mTc – HDP (dose = 9,86MBq/kg). The bone scan revealed high radiopharmaceutical uptake in the myocardium, suggestive for ATTR cardiac amyloidosis. As a result, the patient was referred to undergo genetic testing in order to determine the ATTR subtype involved (wild type - ATTRwt or mutated - ATTRm).

Discussions. Diagnosing ATTR-ca has always represented a problem for the practicing physician. Endomyocardial biopsy is not widely available and presents certain risks. Therefore, a correlation between the existing techniques (biomarker, electrocardiography, ultrasonography) may represent the alternative solution. Nevertheless, these investigations are also limited, due to the fact that they are able to determine the presence of a possible cardiac amyloidosis, but are unable to determine the subtype involved. As a result, more complex investigations are required. Bone scintigraphy has demonstrated a unique ability for detecting and differentiating ATTR-ca from other forms of cardiac amyloidosis that affect the myocardium. The disadvantage in this case is the inability of bone scintigraphy to detect the ATTR subtype involved, therefore additional tests being required (genetic testing). Nevertheless, bone scintigraphy should always be taken into consideration if cardiac amyloidosis is suspected.

Conclusion. If endomyocardial biopsy is not an option and the patient is suspected for cardiac amyloidosis, then bone scintigraphy represents a viable alternative for detecting and differentiating ATTR cardiac amyloidosis. Therefore, this investigation should be considered as a must in the diagnostic algorithm for patients suspected of cardiac amyloidosis.