The sensitivity of DPD scintigraphy to detect transthyretin cardiac amyloidosis in V30M mutation depends on the phenotypic expression of the disease

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Abstract
Background:
There is a growing need for a non-invasive test to detect cardiac involvement in patients with transthyretin-related hereditary amyloidosis (ATTR) caused by V30M mutation. 99mTc-3,3-diphosphono-1,2-propanodicarboxylic acid (DPD) scintigraphy is a promising method, but its accuracy in this particular mutation remains unknown.

Methods:
A cohort of 179 patients: 92 with early-onset disease (EoD, symptoms <50-years-old), 33 with late-onset disease (LoD) and 54 asymptomatic carriers were prospectively evaluated and underwent DPD scintigraphy, which was compared with the results of echocardiogram, ambulatory blood pressure monitoring, 24 h-Holter, myocardial 123I-metaiodobenzylguanidine imaging and NT-proBNP.

Results:
Amyloid cardiomyopathy, defined as septal thickness ≥13 mm, was present in 32 patients (17.9%) and was more frequent in those with LoD (OR: 3.68, p = .003). Cardiac DPD uptake was present in 22 individuals (12.3%) and correlated with parameters indicative of cardiac amyloidosis. DPD imaging was strongly influenced by the age of disease onset: among patients with myocardial thickening, cardiac DPD retention was present in 11/15 (73.3%) with LoD, in contrast to only 4/17 (26.7%) with EoD (p = .005). Two patients with myocardial thickening and normal DPD scintigraphy underwent endomyocardial biopsy that confirmed ATTR amyloidosis.

Conclusion:
DPD scintigraphy presents suboptimal sensitivity to detect cardiac involvement in ATTRV30M, particularly in symptomatic patients with EoD.

Keywords
Amyloidosis, DPD scintigraphy, Scintigraphy, Transthyretin