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CARDIOVASCULAR FLASHLIGHT
*Cardiovascular imaging*

# Multimodality family screening of patients with cardiac transthyretin amyloidosis: a case of an asymptomatic patient

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Transthyretin amyloidosis (ATTR) is a disease caused by amyloid fibrils deposition. There are two types of ATTR, ATTR variant (ATTRv) and ATTR wild-type (ATTRwt). Transthyretin amyloidosis variant is inherited in an autosomal-dominant manner with variable penetrance. Although considered as rare, its prevalence may be underestimated.[1] The disease affects various body systems, most often causing polyneuropathy or cardiomyopathy; however, it may be connected with red flag symptoms such as bilateral carpal tunnel syndrome, biceps tendon rupture, or 'apical sparing' in echocardiography. Without the proper treatment, the disease is associated with short life expectancy, especially when myocardium is affected. This severe, progressive, multi-organ disease carries a poor prognosis with an average survival time of 2–3 years.[1] Transthyretin amyloidosis can be diagnosed based on scintigraphy and the absence of monoclonal protein in serum and urine.[2,3]

A 45-year-old Caucasian, asymptomatic male with a positive family history of ATTRv was admitted to hospital for cardiac evaluation due to a conducted research study [Pfizer Research Grant (ID#57165999) entitled 'The comparisons of regional scintigraphic DPD uptake between patients with hereditary and wild-type cardiac transthyretin amyloidosis']. Laboratory tests showed normal levels of N-terminal-pro-brain natriuretic peptide (NT-proBNP), slightly elevated high-sensitivity troponin levels and the absence of monoclonal protein. Low QRS voltage in limb leads was present in the electrocardiography. The transthoracic echocardiography revealed concentric left ventricle (LV) hypertrophy (mass 291 g, mass index 152 g/cm$^2$, maximal wall thickness 15.5 mm), decreased global LV contractility with LV ejection fraction value of 50%, decreased global LV longitudinal strain (GLS) to −13% with the apical sparing pattern, and first-degree diastolic dysfunction (see Supplementary material online, *Figures S1* and *S2*). Scintigraphy with technetium-99m-3,3-diphosphono-1,2-propanodicarboxylic acid (99mTc-DPD) revealed Perugini 3 myocardial uptake confirmed with single-photon emission computed tomography (SPECT) (*Figure 1*). The genetic test confirmed the pathologic variant, namely, on the transthyretin protein (p.) phenylalanine (Phe) that was replaced by leucine (Leu) (p.Phe53Leu). Therefore, the patient was diagnosed with ATTRv cardiomyopathy and was qualified for the targeted treatment.

In conclusion, screening for ATTR is a crucial part of providing care for the patients' families. In the presented case, noticeable are low levels of NT-proBNP despite the high amyloid burden in the scintigraphy. Multimodality assessment of cardiac amyloidosis is of paramount importance for providing insight into the development of the disease and its current stage. Identifying affected family members may enable early treatment introduction and improvement in their prognosis and quality of life.

## Supplementary material

Supplementary material is available at *European Heart Journal – Case Reports*.

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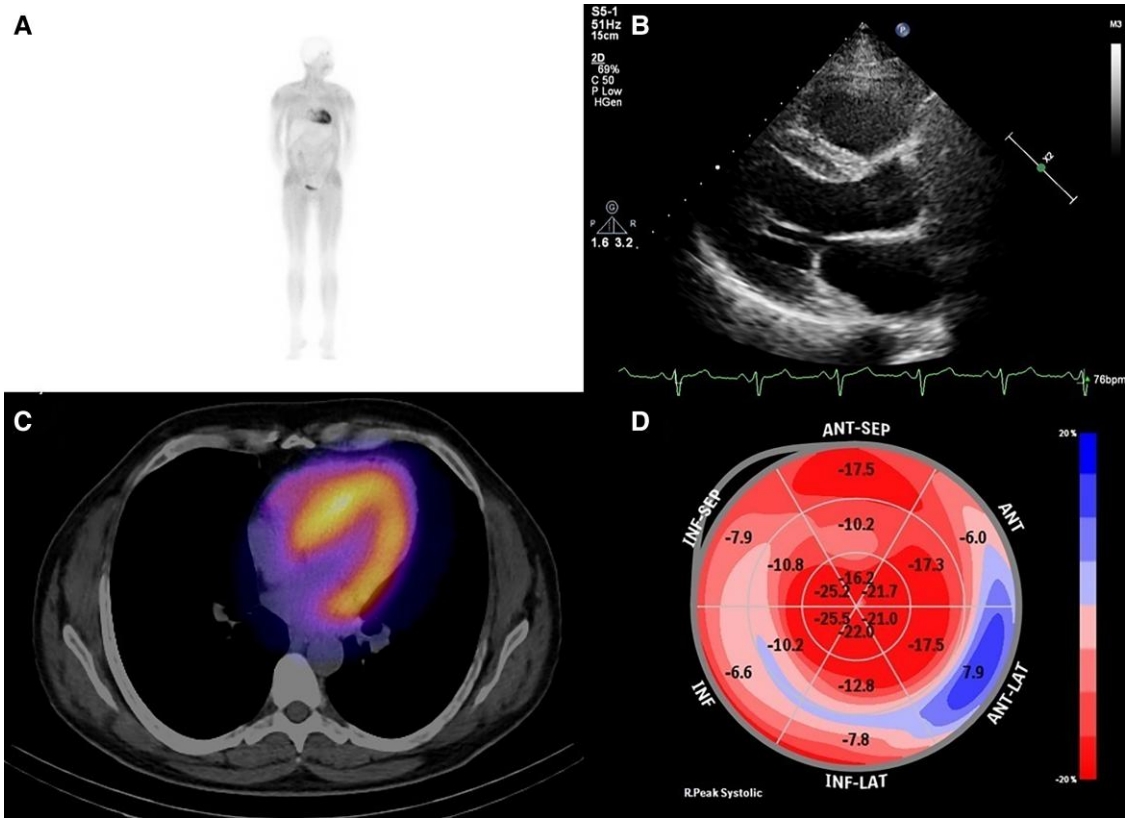


**Figure 1** (*A*) Planar scintigraphic study with administration of technetium-99m-3,3-diphosphono-1,2-propanodicarboxylic acid shows an increased tracer uptake in the vicinity left ventricle consistent with cardiac transthyretin amyloidosis (Perugini 3). (*B*) Parasternal long-axis view, visible classic features of cardiac amyloidosis. (*C*) Hybrid single-photon emission computed tomography with computed tomography showing amyloid deposition in left ventricle region. (*D*) Speckle-tracking imaging, visible preserved apical segmental strain, and reduced basal segmental strain—'apical sparing'.

**Consent:** The authors confirm that written consent for submission and publication of this case report including images and associated text has been obtained from the patient in line with COPE guidance.

**Conflict of interest:** None declared.

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## Data availability

The data underlying this article will be shared on reasonable request to the corresponding author.

## References

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