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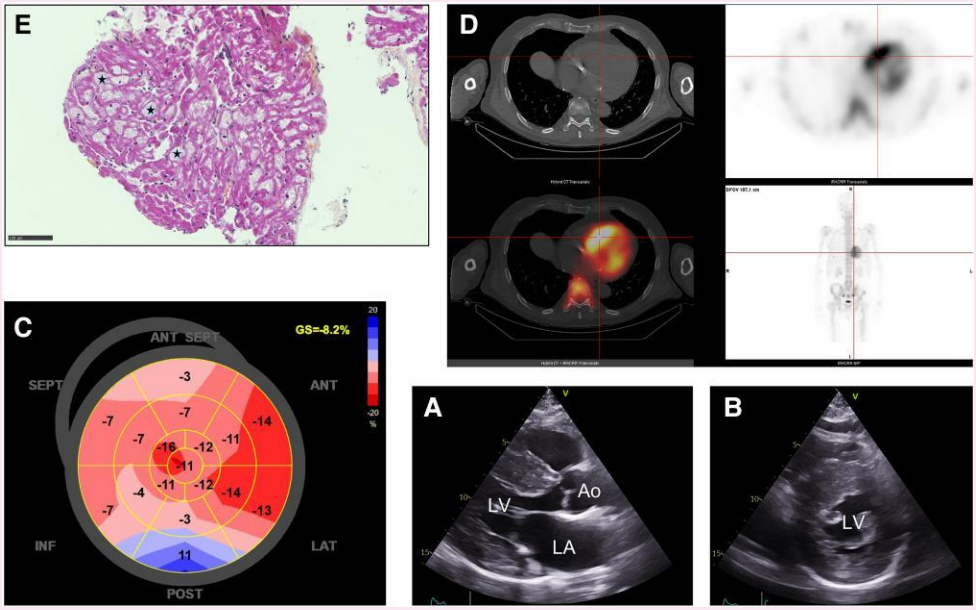
https://doi.org/10.1093/ehjci/jeag072
Online publish-ahead-of-print 11 March 2026

Intense myocardial uptake in a patient with fabry disease—a new cause of false positive of bone scintigraphy mimicking cardiac amyloidosis

Claire Lucas[1]*, Mary Philip[1], Karine Nguyen [2], Victor Morel[2], Laetitia Tessonnier[3], and Gilbert Habib [1]*

[1]Department of Cardiology, CHU La Timone, AP-HM, Aix-Marseille University, 264 rue saint Pierre, Marseille 13005, France; [2]Department of Medical Génétics, CHU La Timone, AP-HM, Aix-Marseille University, Marseille, France; and [3]Department of Nuclear Medicine, CHU La Timone, AP-HM, Aix-Marseille University, Marseille, France

*Corresponding authors. E-mail: claire.lucas@ap-hm.fr; E-mail: gilbert.habib3@gmail.com



Bone scintigraphy has emerged as a key diagnostic tool for transthyretin cardiac amyloidosis (TTR-CA). Despite its high sensitivity and specificity, false positives can occur, causing diagnostic challenges.

A 61-year-old male presented with syncope and left ventricular hypertrophy (LVH) and underwent cardioverter-defibrillator implantation. Genetic testing with a 72-gene panel revealed a pathogenic (class 5) variant in the *GLA* gene: p.(Asn215Ser). He was referred to our cardiomyopathy centre for management of his Fabry disease.

Transthoracic echocardiography showed marked diffuse and symmetrical LVH (*Panels A and B*) (maximal wall thickness = 25 mm), low-normal ejection fraction (55%), and impaired global longitudinal strain (*Panel C*). α-galactosidase A activity was 1.8 µmol/L/h (normal value > 2.8). Migalastat therapy was initiated.

A few months later, the patient underwent bone scintigraphy with 99mTc 3,3-diphosphono-1,2-propanodicarboxylic acid (DPD) coupled with CT imaging for a rheumatologic indication, revealing intense myocardial uptake (Perugini 3), suggestive of TTR-CA (*Panel D*). However, repeated genetic testing excluded *TTR* mutations, and haematologic screening for AL amyloidosis was negative. No known confounding factor associated with false positive bone scintigraphy such as hydroxychloroquine use, recent myocardial infarction, or recent intravenous iron administration was observed. An endomyocardial biopsy found no amyloid deposits but a characteristic vacuolization of cardiomyocytes, confirming Fabry disease-related cardiomyopathy (*Panel E*).

This case underscores a new diagnostic pitfall in interpreting bone scintigraphy, as Fabry disease can mimic TTR-CA by showing intense myocardial uptake. The phenotypic overlap between Fabry disease and CA, including LVH and systemic involvement, can lead to misdiagnosis. A similar case was very recently reported in a patient with Fabry disease, showing myocardial uptake on PYP bone scintigraphy, further supporting the need for clinicians to be cautious when interpreting bone scintigraphy results. Histological confirmation remains crucial in ambigous cases.

Acknowledgements: The authors would like to thank Dr Hubert Lepidi, physician in the Department of Anatomical Pathology at Timone University Hospital, APHM, for the images of the myocardial biopsy.

Author contributions: Claire Lucas (Conceptualization, Data curation, Writing—original draft [lead], Methodology [equal]), Mary Philip (Methodology, Writing—original draft [supporting], Writing—review & editing [equal]), Laetitia Tessonnier (Data curation, Validation [equal], Visualization [supporting]), Karine Nguyen (Writing—review & editing [supporting]), Gilbert Habib (Data curation, Methodology [equal], Writing—original draft [supporting], Writing—review & editing [lead]), and Victor Morel (Investigation, Writing—review & editing [supporting])

Conflict of interest: none declared.

Funding: none declared.

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