



Scatteia, A., De Garate, E., & Bucciarelli-Ducci, C. (2016). 59-year-old female with breathlessness. *Heart*, *102*(20), [1654].
<https://doi.org/10.1136/heartjnl-2016-309836>

Peer reviewed version

License (if available):
CC BY-NC

Link to published version (if available):
[10.1136/heartjnl-2016-309836](https://doi.org/10.1136/heartjnl-2016-309836)

[Link to publication record in Explore Bristol Research](#)
PDF-document

This is the accepted author manuscript (AAM). The final published version (version of record) is available online via BMJ at <http://dx.doi.org/10.1136/heartjnl-2016-309836> . Please refer to any applicable terms of use of the publisher.

University of Bristol - Explore Bristol Research

General rights

This document is made available in accordance with publisher policies. Please cite only the published version using the reference above. Full terms of use are available:
<http://www.bristol.ac.uk/pure/about/ebr-terms>

59-year-old female with breathlessness.

Alessandra Scatteia^{1,2} MD, Estefania De Garate ¹MD, Chiara Bucciarelli-Ducci MD, PHD

1. Bristol Heart Institute, University Hospitals Bristol NHS Foundation Trust, United Kingdom

2. Federico II, University of Naples, Naples, Italy

A 59 year-old female underwent an ECG and echocardiographic screening. Her brother died at a quite young age of kidney failure. Resting ECG showed borderline voltage criteria for left ventricular hypertrophy, with marked widespread T wave inversion. Echocardiogram was normal, but in consideration of exertional breathlessness and abnormal baseline ECG, she underwent a coronary angiogram which showed unobstructed coronaries. She was then referred to have a cardiac magnetic resonance (CMR) for further characterization. CMR images were acquired with a 1.5 Tesla scanner and the imaging protocol included SSFP cine images (Figure 1A) as well as late gadolinium enhancement images (LGE) in the long- and short-axis planes covering the whole left ventricle (Figure 1B). In addition, native and post-contrast T1-mapping (MOLLI) images were acquired for further tissue-characterization (Figure 1C and Figure 1D, respectively).

What is the most likely diagnose, based on CMR findings?

- A. Anderson-Fabry's disease (AFD)
- B. Cardiac amyloidosis
- C. Genotype(+), phenotype(-) hypertrophic cardiomyopathy (HCM)
- D. Myocardial iron overload
- E. Normal heart

Answer A

The wall thickness was normal (Figure 2A). However, this per se does not exclude all the above possible diagnosis, as early phase disease might be present. Post-contrast images showed mid-wall myocardial LGE in the basal inferolateral wall (Figure 2B, white arrow). The native T1 mapping images demonstrate diffusely reduced T1 values, with the exception of the basal inferolateral wall where the values were pseudo-normal (Figure 2C, white arrow).

Given the presence of LGE option E should be excluded, as LGE is not a normal finding.

Option B and C can also be excluded because in both cardiac amyloidosis and HCM the native T1 mapping is normally increased¹⁻³.

Option D can be compatible with low native T1 values but it not typical to find in these patients the LGE and LGE with this pattern.

Post-contrast T1-mapping values were diffusely reduced in all myocardial segments and even lower in the basal and mid-cavity inferolateral wall (Figure 2D arrow), correlating with the presence of LGE. Low native T1-mapping values, together with pseudonormalization of these in the inferolateral wall, have been recently described as patognomonic of AFD^{4,5}.

The diagnosis was confirmed by the detection of low dosage of leucocyte α -galactosidase activity. AFD is an X-linked storage disease caused by deficiency in the enzyme α -galactosidase A. Female patients have been shown to have incomplete phenotypes, most commonly the presence of LGE without LVH⁵. Treatment with recombinant enzyme is available. Our case shows how new CMR relaxometry techniques offer an opportunity for early diagnosis.

References

1. Puntmann VO, Voigt T, Chen Z et al. Native T1 mapping in differentiation of normal myocardium from diffuse disease in hypertrophic and dilated cardiomyopathy. *JACC Cardiovasc Imaging*. 2013 Apr;6(4):475-84;
2. Karamitsos TD, Piechnik SK, Banyersad SM et al. Noncontrast T1 mapping for the diagnosis of cardiac amyloidosis. *JACC Cardiovasc Imaging*. 2013 Apr;6(4):488-97;
3. Germain P, El Ghannudi S, Jeung MY et al. Native T1 mapping of the heart - a pictorial review. *Clin Med Insights Cardiol*. 2014 Dec 1;8(Suppl 4):1-11;
4. Sado DM, White SK, Piechnik SK et al. Identification and Assessment of Anderson-Fabry Disease by Cardiovascular Magnetic Resonance Noncontrast Myocardial T1 Mapping. *Circ Cardiovasc Imaging*. 2013 May 1;6(3):392-8;
5. Pica S, Sado DM, Maestrini V et al. Reproducibility of native myocardial T1 mapping in the assessment of Fabry disease and its role in early detection of cardiac involvement by cardiovascular magnetic resonance. *J Cardiovasc Magn Reson*. 2014 Dec 5;16:99.

This case report presents independent research funded by the National Institute for Health Research (NIHR). The views expressed are those of the author(s) and not necessarily those of the NHS, the NIHR or the Department of Health.

The Corresponding Author has the right to grant on behalf of all authors and does grant on behalf of all authors, an exclusive licence (or non-exclusive for government employees) on a worldwide basis to the BMJ Publishing Group Ltd and its Licensees to permit this article (if accepted) to be published in HEART editions and any other BMJPGl products to exploit all subsidiary rights.