

Native T1 mapping for the diagnosis of Anderson-Fabry disease with myocardial hypertrophy

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A 66-year-old woman previously treated for hypertrophic cardiomyopathy (HCM) underwent cardiac magnetic resonance imaging for further assessment. The modified Look-Locker sequence 5s(3s)3s was performed on a Philips Achieva 3T magnetic resonance scanner (Philips, Amsterdam, Netherlands). Cardiac magnetic resonance had revealed a combined apical and asymmetrical hypertrophy of the left ventricle (Fig 1). There was a diffuse, non-ischaemic pattern of late gadolinium enhancement in the region of hypertrophy in keeping with HCM. The septal:lateral wall ratio on the 4-chamber view was 2.2:1. The mid-inferoseptal:anterolateral ratio on the short-axis view was 1.32:1. However, native T1 mapping was abnormally low (1009 ms; normal range on our scanner, 1226-1256 ms) [Fig 2]. T2 mapping values were in the normal range indicating no underlying iron deposition to account for the low T1 values. Low native T1 mapping values are atypical of HCM that would normally be associated with a slight increase in native T1. This suggested underlying Anderson-Fabry disease (AFD), subsequently proven by genetic testing in this patient.

Anderson-Fabry disease is an uncommon X-linked sphingolipid storage disorder resulting

from deficiency of the lysosomal enzyme α -galactosidase. The principal driver of mortality in AFD is cardiac disease.¹ Disease manifestations include left ventricle hypertrophy that can mimic HCM.² Other complications of AFD include valve thickening, myocardial scarring, cardiac failure and arrhythmic death.³ Enzyme replacement therapy for AFD should be started early to prevent progression of cardiac disease. Cardiac magnetic resonance myocardial native T1 is decreased in AFD and is a non-invasive method that may raise suspicion of AFD in the context of left ventricle hypertrophy or suspected HCM. Native T1 mapping is an established reproducible technique.¹ Raising suspicion and later confirming AFD is crucial to managing these patients appropriately as well as initiating screening in asymptomatic family members.

Author contributions

All authors have made substantial contributions to the concept or design of this study; acquisition of data; analysis or interpretation of data; drafting of the manuscript; and critical revision for important intellectual content. All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

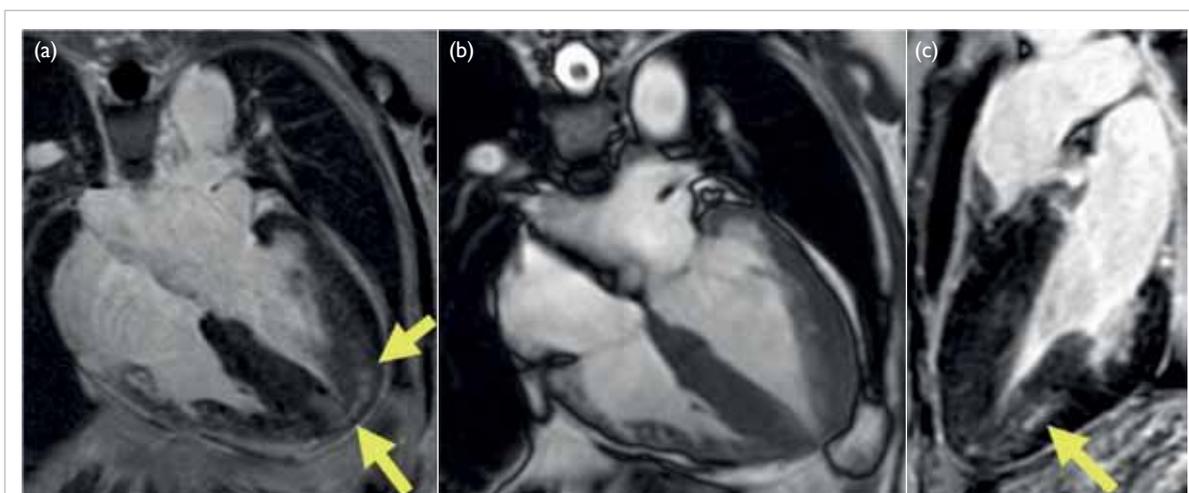


FIG 1. Cardiac magnetic resonance imaging of the patient demonstrating the following: (a) 4-chamber late gadolinium enhancement (LGE) image showing diffuse mid-wall LGE at the mid-ventricular and apical left ventricular walls (arrows); (b) 4-chamber cine image showing hypertrophy of the lateral and septal walls with a septal:lateral wall ratio of 2.2:1; and (c) 2-chamber LGE image demonstrating mid-wall LGE particularly in the inferior apical wall (arrow)

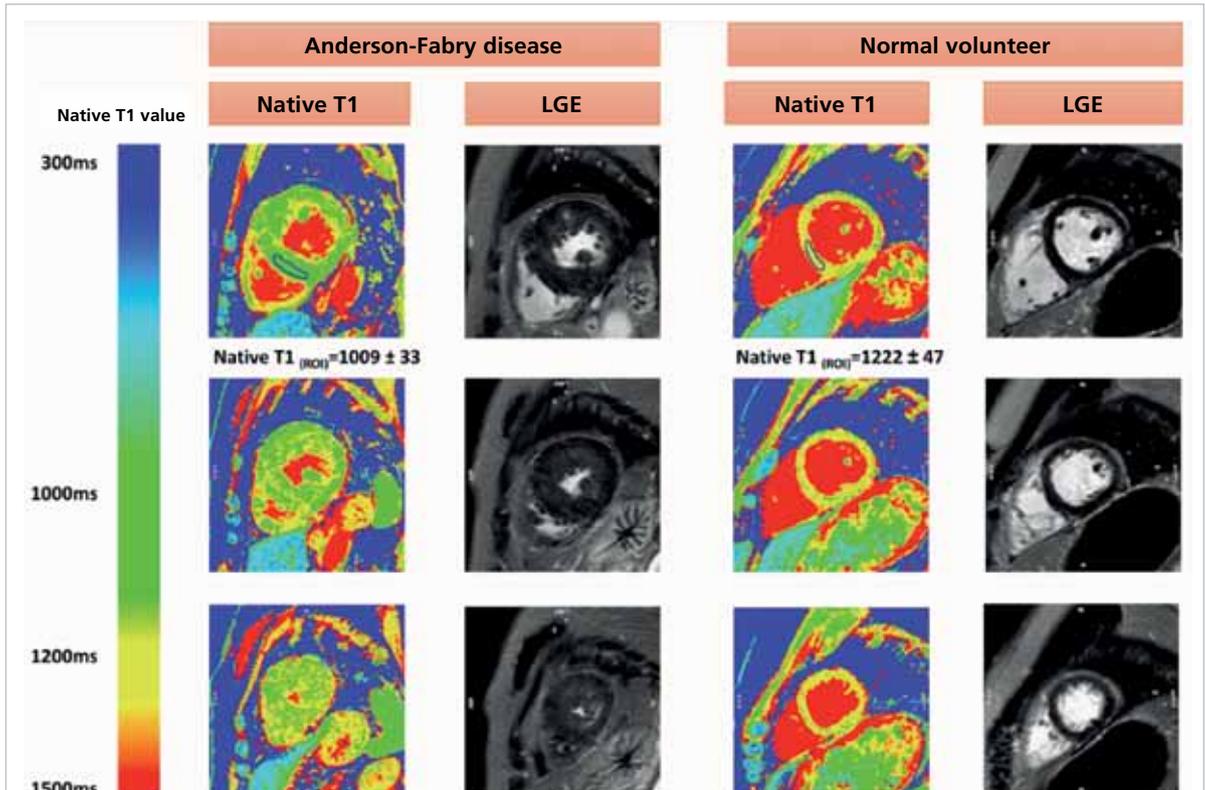


FIG 2. Native T1 mapping images comparing an Anderson-Fabry's disease (AFD) patient with cardiac involvement (left-sided images) and a normal volunteer (right-sided images). Abnormally reduced T1 values are seen in the AFD patient despite a diffuse, non-ischaemic pattern of late gadolinium enhancement (LGE) that would usually result in elevation of T1 values (native T1 mapping of the AFD patient region of interest (ROI) = 1009 ± 33 ms compared with the normal volunteer [ROI = 1222 ± 47 ms]). The ROI was measured at the basal septal wall. The native T1 values at the mid (1161 ms) and apical short axis (1135 ms) views were also reduced

Conflicts of interest

The authors have no conflicts of interest to disclose.

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Ethics approval

Relevant patient consent was obtained for the purpose of this case study.

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