Rare myeloid neoplasm with an abnormal left ventricular mass: impact of cardiovascular magnetic resonance tissue characterisation for diagnosis and guiding management

Francesco Papalia ¹, Sara Tyebally,^{1,2} Abbas Zaidi,³ Mohammed Khanji ^{1,4,5}

¹Cardiology, Barts Heart Centre, Barts Health NHS Trust, London, UK

²Hatter Cardiovascular Institute, University College London, London, UK

³Haematology, Barking Havering and Redbridge Hospitals NHS Trust, Romford, UK

⁴Cardiology, Newham University Hospital, Barts Health NHS Trust, London, UK

⁵NIHR Barts Biomedical Research Centre, William Harvey Research Institute, Queen Mary University of London, London, UK

Correspondence to
Dr Mohammed Khanji;
m.khanji@qmul.ac.uk

Accepted 6 February 2022

DESCRIPTION

A man in his 40s presented with progressive peripheral oedema, troponin T 14 ng/L (normal range <14 ng/L) and elevated eosinophils $12.9 \times 10^9/L$ (normal range $0-0.4 \times 10^9/L$). Echocardiography revealed abnormal left ventricular apical thickening. An initial diagnosis of systemic mastocytosis was made, but subsequent bone marrow aspirate identified a FIP1L1-PDGFR gene fusion, confirming the diagnosis of myeloid neoplasm with PDGFR rearrangement.

Cardiovascular magnetic resonance (CMR) (figure 1) revealed low normal left ventricular (LV) systolic function (ejection fraction 59%) with apical LV cavity obliteration (figure 1A see also video 1). Apical myocardial T1 was elevated (figure 1B, apical septal = 1366 ms, apical lateral = 1377 ms, normal = 1220–1360 ms) but normal T2, excluding oedema/inflammation. Apical hypointensity on early gadolinium enhancement (arrow, figure 1C) and extensive mid-apical late gadolinium enhancement (figure 1D) suggested eosinophilic endomyocardial fibrosis (Loeffler's

Video 1 Cine sequence demonstrating mildly impaired systolic function and the apical LV mass throughout the cardiac cycle.

Figure 1 Showing the apical LV cavity mass (A), raised T1 mapping values in the apical myocardium (B), hypointensity on early gadolinium enhancement due to extensive LV thrombus (C), and pronounced apical endomyocardial fibrosis on late gadolinium enhancement (D).

syndrome) complicated by a large thrombus requiring anticoagulation.

The FIP1L1-PDGFR syndrome is rare and cardiac involvement is not always present. The phenotype is variable and some patients are asymptomatic though others can have multi-system involvement due to eosinophilic tissue infiltration.

The condition is highly sensitive to imatinib which can limit the degree of myocardial fibrosis; however, detection of the causative rearrangement is specialised and can be missed on initial sequencing by gene panels, as in this

Learning points

- ▶ FIP1L1-PDGFR mutation is rare in myeloid neoplasms but the association with cardiac involvement has been well described. Cardiac involvement in systemic mastocytosis is much more infrequent.
- ▶ Cardiovascular magnetic resonance imaging is the gold standard modality to confirm the extent of cardiac involvement and quantify thrombus burden, enabling timely appropriate management.

© BMJ Publishing Group Limited 2022. No commercial re-use. See rights and permissions. Published by BMJ.

To cite: Papalia F, Tyebally S, Zaidi A, et al. *BMJ Case Rep* 2022;**15**:e245312. doi:10.1136/bcr-2021-245312

case. Early recognition of cardiac involvement is important as ventricular dysfunction may become irreversible, although routine screening for cardiac complications is not currently recommended. Cardiac management is based around guideline-directed medical therapy of LV dysfunction if present, alongside definitive management of the underlying neoplasm. Multimodality imaging, including CMR, are key to diagnosis and guiding optimum management.

Contributors FP, ST and MYK supervised scanning and interpreted the cardiac MRI. FP, ST, AZ and MYK contributed to the writing, revision and decision to submit the case report.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent for publication Consent obtained directly from patient(s).

Provenance and peer review Not commissioned; externally peer reviewed.

Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

ORCID iDs

Francesco Papalia <http://orcid.org/0000-0002-1059-0169>
Mohammed Khanji <http://orcid.org/0000-0002-5903-4454>

Copyright 2022 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <https://www.bmj.com/company/products-services/rights-and-licensing/permissions/>
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

Customer Service

If you have any further queries about your subscription, please contact our customer services team on +44 (0) 207111 1105 or via email at support@bmj.com.

Visit casereports.bmj.com for more articles like this and to become a Fellow