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

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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 × 10^9/L (normal range 0–0.4 × 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-PDGFRA gene fusion, confirming the diagnosis of myeloid neoplasm with PDGFRA 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 syndrome) complicated by a large thrombus requiring anticoagulation.

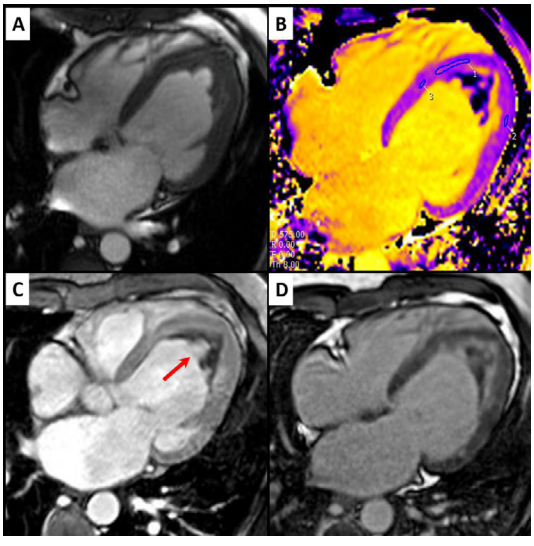
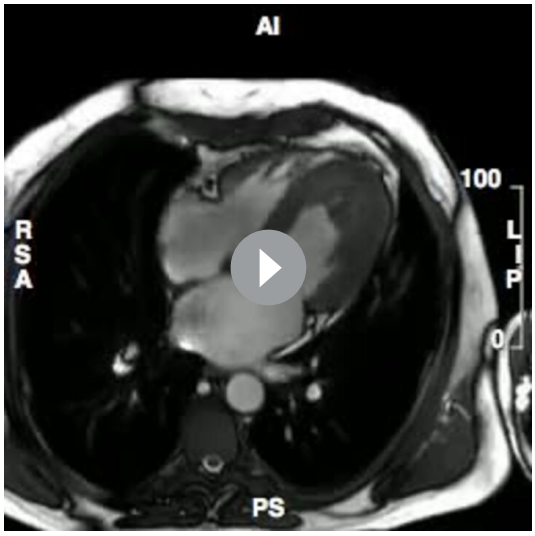


Figure 1 Showing the apical LV cavity mass (A), raised T1 mapping values in the apical myocardium (B), hypoattenuation on early gadolinium enhancement due to extensive LV thrombus (C), and pronounced apical endomyocardial fibrosis on late gadolinium enhancement (D).



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

The FIP1L1-PDGFRA syndrome is rare and cardiac involvement is not always present. The phenotype is variable and some patients are asymptomatic though others can have multisystem 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-PDGFRA 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.

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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.

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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.

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