

## Rare disease

## Endomyocardial fibrosis causing stroke in a young man

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## Summary

An Indian man in his late 30s presented with ischaemic stroke and eosinophilia of 711/mm<sup>3</sup>. ECG showed first-degree heart block with ST depression and symmetrical T-wave inversions in the chest leads. Subsequently, the patient was further evaluated by echocardiography and cardiac MRI which identified the presence of endomyocardial fibrosis in the heart.

## BACKGROUND

Endomyocardial fibrosis is a very rare cause of stroke, which is even more unusual in the absence of hypereosinophilia.

## CASE PRESENTATION

A 38-year-old South-Indian immigrant presented with a history of dizziness, left-sided body weakness and slurred speech that was noted upon waking up from sleep. He had been experiencing mild dyspnoea on exertion for the past few months. His medical history was unremarkable except for mild dyspnoea on exertion without orthopnea or paroxysmal nocturnal dyspnoea. There were no preceding symptoms of chest pain, palpitation, weight loss, fever or rash. He was not known to be asthmatic, diabetic or hypertensive. Family history was unremarkable. He smokes cigarette occasionally.

On initial examination, blood pressure was 132/90 mm Hg, pulse was 74/min and regular. Neurological examination revealed decreased tone and power (3/5) with extensor planter reflex on the left side. There was also left-sided upper motor neuron lesion (UMNL) of the facial nerve. Other systemic examination was unremarkable.

## INVESTIGATIONS

Biochemical and haematological investigations were within normal range apart from a raised eosinophil count of 711/mm<sup>3</sup> (upper limit of normal 400/mm<sup>3</sup>) and raised erythrocyte sedimentation rate of 30 mm in first hour. Autoimmune profile and thrombophilia profile were negative. Stool testing for ova and parasite was also negative.

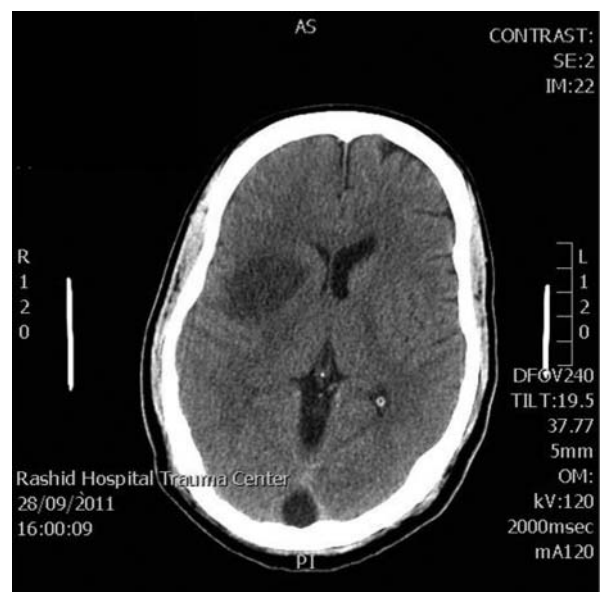
Brain CT showed evidence of right basal ganglia infarction (figure 1). Carotid Doppler study was normal.

ECG showed sinus rhythm, first-degree atrioventricular (AV) block, ST segment depression in leads V4–V6, I and aVL and T-wave inversion in leads V2–V6, II and aVL (name given for augmented left limb lead in ECG) (figure 2). Subsequently, the patient was referred to the cardiology team for further evaluation.

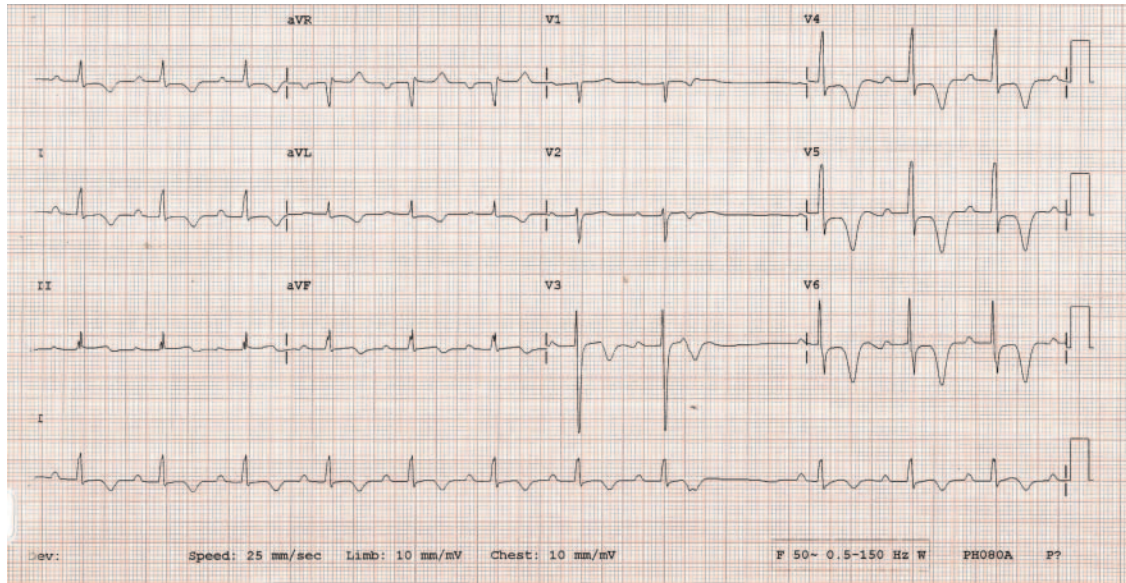
Transoesophageal echocardiography showed thickening of the right ventricular (RV) and left ventricular (LV) apices with prominent calcification at endocardial surface in LV, causing obliteration of both the ventricular cavities (figure 3). LV systolic function was impaired with an LV ejection

fraction of 35%. There was mild mitral regurgitation and both atria were of normal dimensions. There is enhanced density of the moderator band of the RV apex. There was no evidence of pericardial effusion or intracardiac thrombi. Then patient underwent right and left heart catheterisation which demonstrated normal coronary arteries and the ventriculogram demonstrated obliteration of both ventricles with impaired systolic function (figure 4).

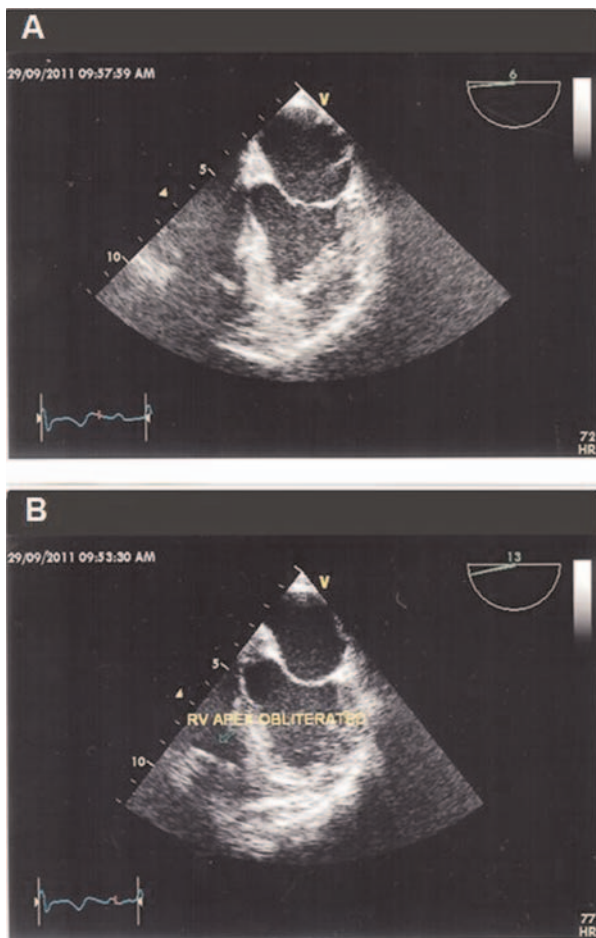
Finally, cardiac MRI was done, which showed myocardial thickening involving biventricular apex, inter-ventricular septum extending to mid-cavity causing obliteration of mid- and apical cavity (figures 5 and 6). The lesion was isointense to the rest of the myocardium with focal areas of calcification. LV apex and all four apical segments were akinetic. With delayed enhancement, focal mid-myocardial and subepicardial enhancement was seen at both ventricular apices (figures 7 and 8). Collectively,



**Figure 1** Brain CT showed a hypodense area in the right basal ganglia with effacement of cortical sulci in the right frontal area and compression of ipsilateral lateral ventricle.



**Figure 2** ECG showed sinus rhythm, first-degree atrioventricular block, ST segment depression in leads V4–V6, I and aVL and T-wave inversion in leads V2–V6, II and aVL.

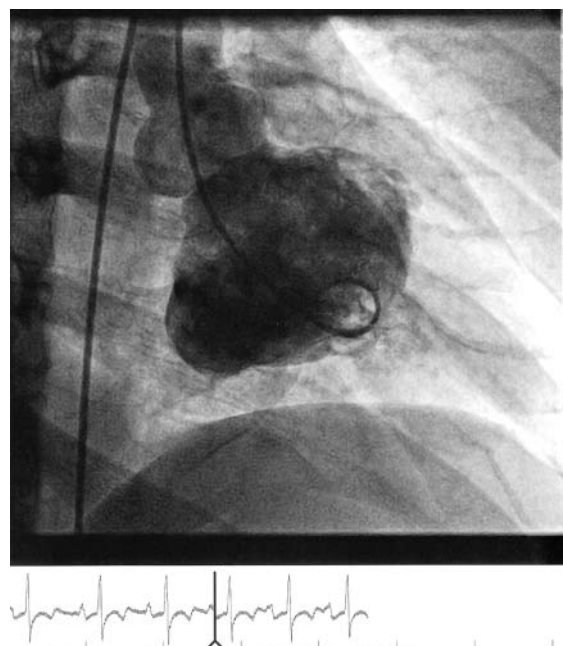


**Figure 3** Echocardiography showed obliteration of left ventricular (A) and right ventricular apices (B).

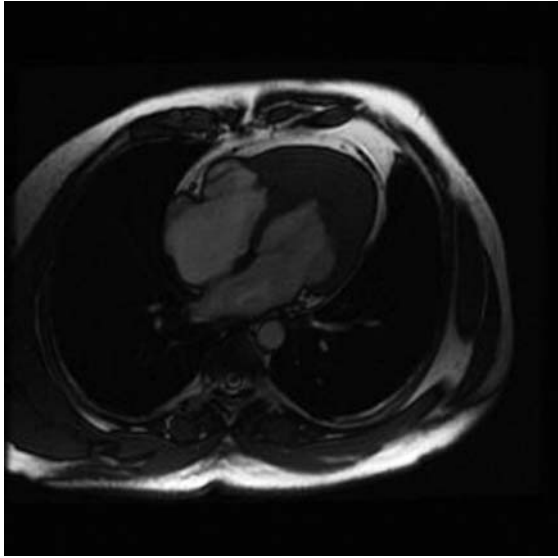
these findings were highly suggestive for endomyocardial fibrosis (EMF).

**DIFFERENTIAL DIAGNOSIS**

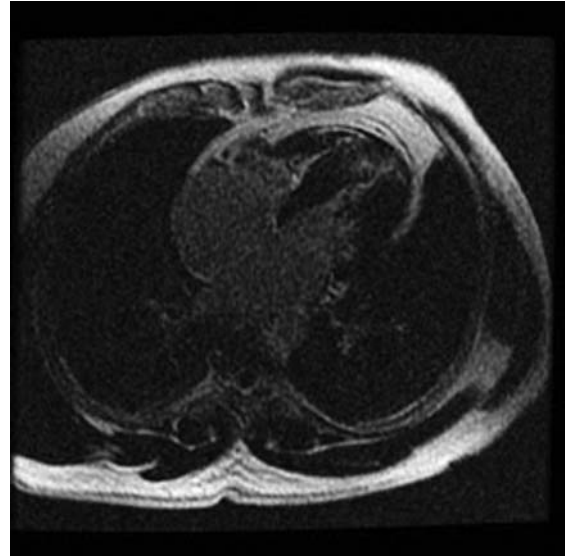
- ▶ Hypertrophic cardiomyopathy
- ▶ Apical clot
- ▶ Amyloidosis



**Figure 4** Left ventricular ventriculogram revealed obliteration of the left ventricular cavity.



**Figure 5** T1-weighted MRI image of the heart showing obliteration of right and left ventricle apices.



**Figure 7** With delayed enhancement, focal mid-myocardial and subepicardial enhancement was seen at both ventricular apices.

**TREATMENT**

The patient was initiated on warfarin for anticoagulation with a target international normalized ratio of 2–3. He was also referred to the physiotherapy department for stroke rehabilitation. The course of his stay in the hospital was uneventful.

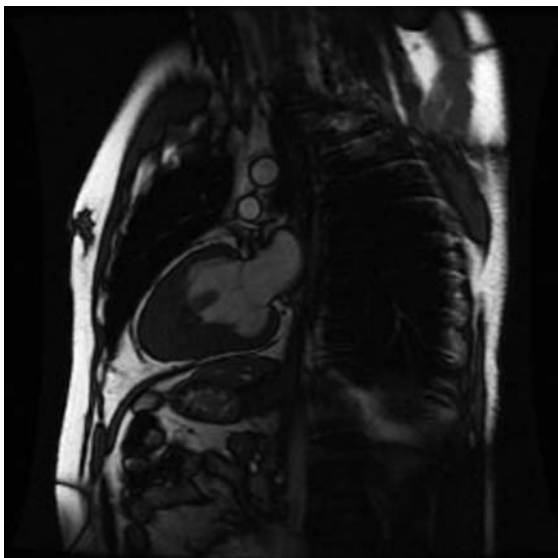
**OUTCOME AND FOLLOW-UP**

Patient came for a follow-up visit after 3 months. His neurological status had improved considerably. He had mild residual weakness on the left side of his body, but was otherwise able to return to his former level of functional capacity.

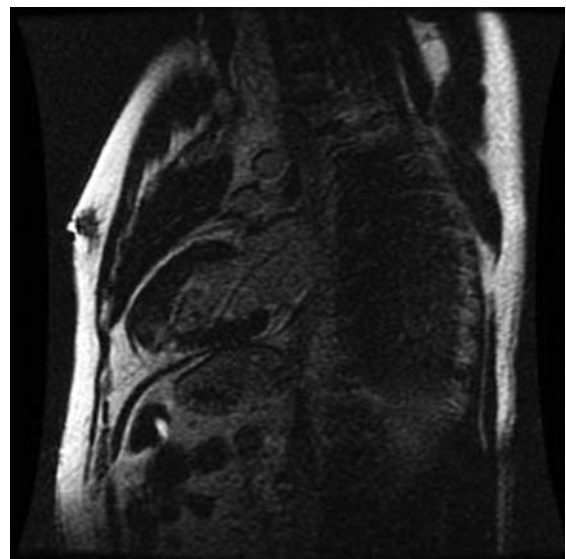
**DISCUSSION**

Several cardiac conditions predispose to the occurrence of cardio-embolic strokes, among which EMF is a relatively uncommon cause.

EMF was first identified by the pathologist, Jack NP Davies in Uganda in 1947. Today, it is recognised as the most common form of restrictive cardiomyopathy worldwide,<sup>1</sup> which is mainly prevalent in the tropical countries such as certain parts of South America,<sup>2</sup> sub-Saharan Africa<sup>3 4</sup> and India.<sup>5</sup> A study conducted in the rural areas of Mozambique found the overall prevalence of EMF to be 19.8%, with the highest prevalence in persons 10–19 years of age.<sup>6</sup>



**Figure 6** T1-weighted MRI image of the heart showing obliteration of left ventricular apex.



**Figure 8** Cardiac MRI with in sagittal view showed subendocardial and myocardial delayed enhancement in ventricular apex.

EMF starts as fibrosis of the apical endocardial surface which progressively causes increased stiffness and reduction in cavity of the involved ventricle. This ultimately results in restrictive physiology. The exact aetiology for this disease still remains to be unknown; however, certain triggering factors such as geography, unidentified infective agents and eosinophilia have been postulated to be involved.<sup>7-9</sup>

Since the disease process progresses insidiously, the initial symptoms can be very subtle—as in our patient who had a preceding history of dyspnoea on exertion. A study by Mocumbi *et al*<sup>6</sup> showed that only 22.7% of those who had EMF were symptomatic at time of detection. The clinical manifestation of this disease is predominantly related to symptoms and signs of heart failure. Thromboembolic events are an uncommon initial presentation of this disease. Several case studies in the literature have highlighted the occurrence of systemic thromboembolic events in patients with hypereosinophilic syndrome.<sup>10-14</sup> However, our patient had developed ischaemic stroke with elevated eosinophil count that did not reach the range of hypereosinophilia.

A study by Seth *et al*<sup>5</sup> reported that majority of patients with EMF had involvement of both ventricles. Mocumbi *et al*<sup>6</sup> proposed diagnostic criteria for EMF based on echocardiographic features. The disease is diagnosed in the presence of two major criteria or one major criterion associated with two minor criteria (table 1). Our patient's echo findings included endomyocardial plaques, obliteration of LV and RV apices, and enhanced density of the moderator band. He thus fulfilled the criteria for echocardiographic diagnosis of EMF.

Cardiac MRI is increasingly receiving increasing importance as a diagnostic tool in the evaluation of patients with heart failure.<sup>15 16</sup> Recent studies have highlighted the role of late gadolinium enhancement MRI in

identifying the characteristic endomyocardial fibrous tissue deposition which is the hallmark of EMF.<sup>17</sup>

There are no well-designed randomized controlled trials that have assessed the role of medical intervention in EMF. The medical treatment revolves around management of heart failure.<sup>1</sup> However, selected patients may benefit from surgical interventions, such as endocardial resection with atrioventricular valve replacement<sup>18</sup> or heart transplantation.<sup>19</sup>

EMF has a poor prognosis because of the natural course of disease, as it leads to complications such as heart failure, arrhythmias and systemic thromboembolism. It is an underdiagnosed cause of cardiomyopathy in the endemic regions.<sup>20</sup> And when it occurs outside the endemic region, it poses an even greater diagnostic challenge for physicians, as it clinically mimics many other causes of cardiomyopathy.<sup>21</sup>

**Learning points**

- ▶ Cases of endomyocardial fibrosis (EMF) are increasingly detected in non-endemic regions due to influx of immigrants from endemic regions.
- ▶ Consider a diagnosis of EMF in young patients from endemic regions who present with thromboembolism and eosinophilia.
- ▶ Consider a diagnosis of EMF in a young patient who develops heart failure.
- ▶ Cardiac MRI is an important tool that can be used for evaluation of patients with restrictive cardiomyopathy.

**Competing interests** None.

**Patient consent** Obtained.

**Table 1** Echocardiographic diagnostic criteria for EMF

| Criterion  | Score  |
|--|--|
| <b>Major criteria</b>  |  |
| Endomyocardial plaques >2 mm in thickness  | 2  |
| Thin (≤1 mm) endomyocardial patches affecting more than one ventricular wall                         | 3  |
| Obliteration of the right ventricular or left ventricular apex                                       | 4  |
| Thrombi or spontaneous contrast without severe ventricular dysfunction                               | 4  |
| Retraction of the right ventricular apex (right ventricular apical notch)                            | 4  |
| Atrioventricular valve dysfunction due to adhesion of the valvular apparatus to the ventricular wall | 1-4 (depending on the severity of the regurgitation) |
| <b>Minor criteria</b>  |  |
| Thin endomyocardial patches localised to 1 ventricular wall  | 1  |
| Restrictive flow pattern across mitral or tricuspid valves   | 2  |
| Pulmonary-valve diastolic opening  | 2  |
| Diffuse thickening of the anterior mitral leaflet  | 1  |
| Enlarged atrium with normal size ventricle   | 2  |
| M-movement of the interventricular septum and flat posterior wall                                    | 1  |
| Enhanced density of the moderator or other intraventricular bands                                    | 1  |

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