International Journal of Scientific Reports
Bassey K et al. Int J Sci Rep. 2022 Jun;8(6):151-154
http://www.sci-rep.com
pISSN 2454-2156 | eISSN 2454-2164

DOI: https://dx.doi.org/10.18203/issn.2454-2156.IntJSciRep20221340

Case Report

Endomyocardial fibrosis in a 7-year-old Nigerian child: a case report and review of the literature

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Received: 22 April 2022
Revised: 16 May 2022
Accepted: 17 May 2022

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ABSTRACT

Endomyocardial fibrosis (EMF) is a poorly-understood idiopathic disorder characterized by the development of restrictive cardiomyopathy. It is a disorder of the tropics and subtropics and was once the second commonest cause of acquired heart disease in Nigeria. Its diagnosis is often made when the disease is advanced and the prognosis is invariably poor at this stage. Reports of EMF have become increasingly rare in recent years and it is frequently misdiagnosed. This report seeks to highlight EMF as a still present and potentially important cause of heart failure in Nigerian children. A 7-year-old male referred to the pediatric cardiology clinic with complaints of abdominal and facial swelling, with no difficulty in breathing and exercise intolerance. Examination revealed a classic "egg-on-stick" appearance. Apex was not displaced and heart sounds were distant. Liver was enlarged but non tender and there was demonstrable ascites. A 2D echocardiography showed massive right atrial enlargement with severe tricuspid regurgitation and fibrosis within the right ventricular cavity which was small. There was a moderate-sized pericardial effusion. Electrocardiogram showed right atrial enlargement with low-voltage complexes. Full blood count showed eosinophilia (20% total white blood cell count). Total serum protein and albumin were reduced. EMF, though increasingly rare, remains an important cause of childhood cardiac morbidity and mortality. A high index of suspicion is critical as the prognosis remains poor.

Keywords: Endomyocardial fibrosis, Restrictive cardiomyopathy, Heart failure, Nigerian children, Tropics

INTRODUCTION

Endomyocardial fibrosis (EMF) is an idiopathic disorder characterized by the development of restrictive cardiomyopathy and fibrotic changes of the endocardial lining and in particular, the cardiac apex.[1] It is the commonest cause of restrictive cardiomyopathy worldwide. It was first described in 1948 by Davies in Uganda but its etiology and pathogenesis are still poorly elucidated and remains enigmatic.[2] Eosinophilia, chronic parasitic infestation as well as nutritional factors such as high-tuber diets, cerium toxicity and hypomagnesaemia have been suggested as possible etiologies.[3] No specific treatment is known. It is primarily a disease of the tropical and sub-tropical region of the world but isolated cases have been reported in temperate regions.[4] EMF is considered by many to be part of a spectrum of disorders which include tropical EMF and Loeffler endocarditis (non-tropical eosinophilic EMF).[5] Both conditions have also been known to present concurrently, on rare occasions.[5] The clinical course of EMF is marked by late presentation in patients who already have advanced cardiac lesions and resulting poor prognosis.

The prevalence of EMF is highest in the low-income communities of the rainforests of Africa, India and South America which are within 15° of the equator. There have been reports published from the drier Savannah region of Northern Nigeria, however EMF is very rarely reported in persons who have not visited the tropics or sub-tropics.[4,6] EMF remains a very common cause of cardiac disease in certain parts of East Africa and changes

consistent with EMF were found in about 20% of a random sample of subjects of all ages in Mozambique.[7] In Nigeria, EMF accounted for up to 10% of all cardiac diseases in the 1960s and 1970s and was the second commonest cause of acquired heart disease in the country.[8] The prevalence of EMF has however declined significantly in the last two decades and now accounts for between 0.04 and 0.09% of all cardiac cases.[9,10] A similar decline in prevalence rates has also been noted in India.[11] In a recent survey of 1441 cardiac patients referred for echocardiography in South Western Nigeria, there was no diagnosis of EMF made.[12]

Despite this apparent reduction in incidence and the relative absence of pediatric cases in large reviews, case reports of children with EMF continue to be published and such children are often misdiagnosed as having other conditions such as tuberculosis or Ebstein anomaly.[12-16] The frequent late presentations, poor prognosis and the possibility of misdiagnosis underscore the need for a high index of suspicion to be maintained in the face of declining cases to ensure that the best possible outcomes are achieved for children who present with EMF.

This report seeks to highlight the fact that EMF is still an important cause of heart failure in the Nigerian child. It also seeks to highlight the potential for misdiagnosis.

CASE REPORT

A 7-year-old male was referred to the pediatric cardiology clinic with complaints of abdominal and facial swelling of three months duration, with no difficulty in breathing or exercise intolerance. There was associated early satiety but no vomiting or abdominal pain. There were no previous admissions or surgeries. Child frequently ingested water-based herbal concoctions.

Examination revealed a chronically ill looking school-aged child in no obvious respiratory distress with bilateral parotid fullness. He had a classic "egg-on-stick" appearance (Figure 1).

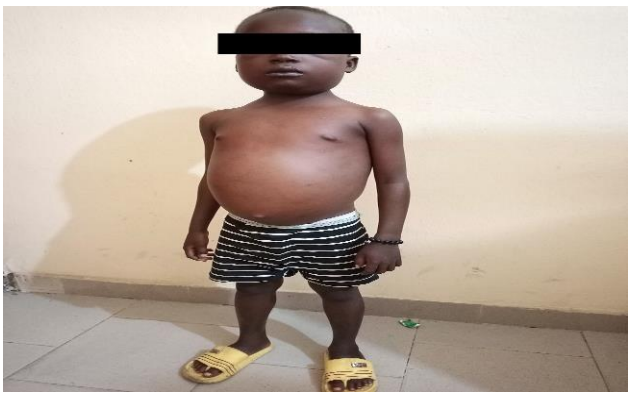

Figure 1: Image of patient showing "egg-on-stick" appearance.
The image demonstrates the marked abdominal swelling with absent pedal edema in the index patient.

He was mildly pale, anicteric, with no pedal oedema or peripheral lymph node enlargement. His weight was 18.1 kg (-2.2Z). Height-110 cm (-2.5Z). His pulse rate was 86 bpm and the pulse was regular and moderate volume. BP was 110/70 mmHg. JVP was raised. Apex was not displaced but was diffuse. No thrills. S1 and S2 only were heard. Heart sounds were distant. Abdomen was distended; liver was enlarged 4 cm but not tender and there was ascites. Respiratory rate was 28 cpm, he was not dyspneic. Percussion notes were dull on the left hemithorax. There vesicular breath sounds bilaterally.

The differential diagnosis included endomyocardial fibrosis and chronic liver disease.

The full blood count showed eosinophilia (20% total WBC count). The WBC count 5.9×10^9/l. His PCV 35%.

Total serum protein was 47 g/l (62-82) and albumin was 26 g/l (36-52).

Chest X-ray showed normal cardiac size and silhouette shape.

A 2D echo showed massive right atrial enlargement within which was spontaneous contrast; there was also fibrosis within the right ventricular cavity which was reduced in size (Figure 2). There was tricuspid regurgitation, however no thrombus was demonstrated. There was systolic dysfunction of the right ventricle (Figure 3) with paradoxical septal motion. There was a moderate-sized pericardial effusion.

Electrocardiogram showed right atrial enlargement with low-voltage complexes and qR pattern in V1 (Figure 4). There was no evidence of AV block or atrial fibrillation.

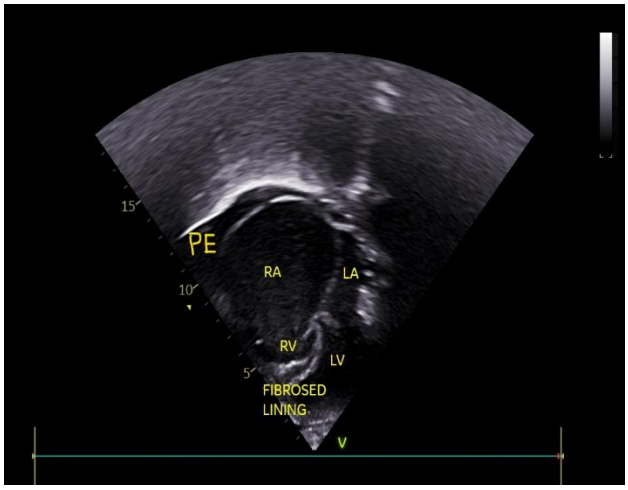

Figure 2: 2D Echo Image demonstrating features of EMF.
This image demonstrates the salient features of EMF with right atrial dilatation, reduced RV chamber size and fibrosis of the RV. The pericardial effusion is also seen around the RA. RA: right atrium, RV: right ventricle, PE: pericardial effusion, LA: left atrium, LV: left ventricle.

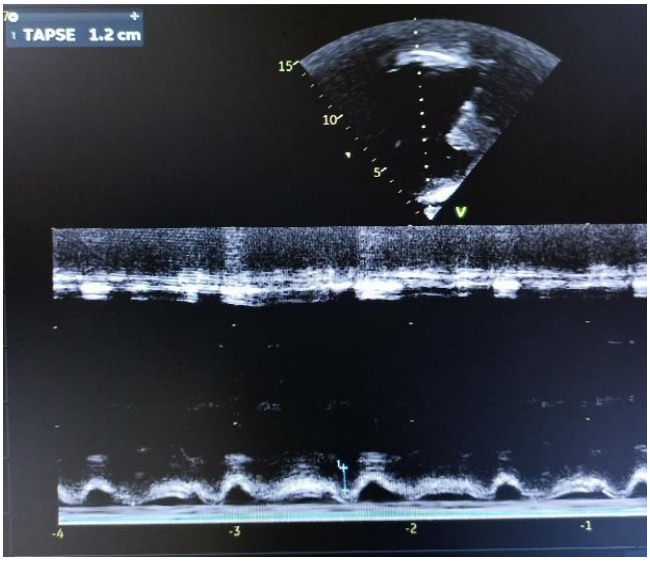

Figure 3: 2D echo image demonstrating TAPSE measurement.

This image demonstrates the TAPSE measurement. The value was -4.9Z for our patient.

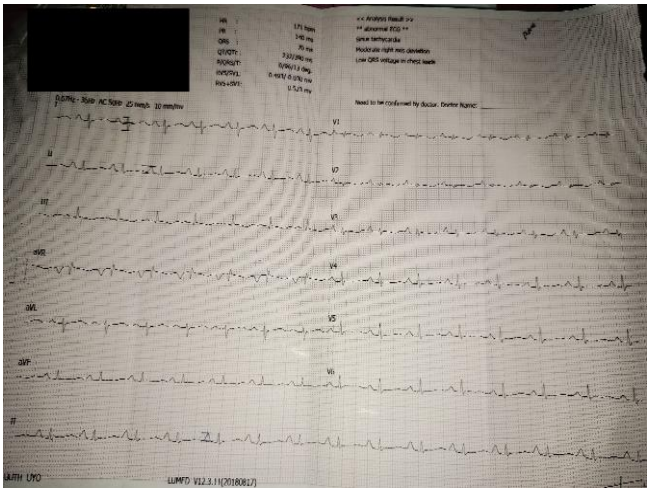

Figure 4: Electrocardiogram of patient.

This is an image of the patient's electrocardiogram. Low voltage QRS complexes can be seen. There are also peaked P waves demonstrating right atrial enlargement and a qR pattern may be seen in V1.

He was treated with furosemide, hydrochlorothiazide and captopril. He has had four follow up visits and has remained stable.

DISCUSSION

Endomyocardial fibrosis, although it has become rare in our environment, remains an important cause of pediatric cardiac morbidity in our environment. Most patients present when the disease is well into the fibrotic stage and have poor outcomes. There is also a potential for diagnostic delays due to misdiagnosis even after echocardiography. Our initial differential diagnosis for the patient included Ebstein anomaly due to the reduced RV size and the relative inferior displacement of the tricuspid valve. This has also been previously reported.[15]

The index patient presented with predominantly right ventricular EMF (RV EMF) which is the commoner form of univentricular EMF. Biventricular disease is however the most common form of the disease, accounting for 55% of all cases.[17] The finding is however consistent with recent reports of predominantly RV EMF including a case series in Sudanese children which showed isolated RV EMF in five out of six cases and biventricular EMF in the last case, a case series from South Eastern Nigeria involving patients of all ages.[10,15] In a case series of children from Malawi, RV EMF was identified in all cases.[14] Other recent case reports have also identified RV EMF in children.[13,16] The clinical features of abdominal distension, undernutrition with no pedal oedema and the absence of dyspnea and easy fatiguability are also in keeping with a diagnosis of RV EMF.[17-18] The paradoxical absence of significant peripheral oedema despite the marked right ventricular dysfunction in our index patient is a feature that is classical of EMF. The finding is poorly understood.[14]

The age of the index patient (seven years) is well within the peak age range of childhood EMF.[18] EMF is rarely reported before the age of four years although recent reports have shown that the diagnosis can be made as early as during the neonatal period.[13,16] There is no established sex predilection for EMF.[18] Recent reports from Nigeria have suggested a slight male preponderance, and in a study from Sudan, all identified children with EMF were male.[9,10,15] The M:F ratio in a Malawian study was 1:1.[14]

The echocardiographic features were characteristic of EMF with four out of five prime diagnostic criteria being present in the index patient viz; ventricular wall fibrosis, huge atrium, atrioventricular valve regurgitation and obliteration of ventricular cavity.[19] The presence of a pericardial effusion in the patient lends further diagnostic support. The electrocardiographic findings of low QRS voltages and atrial enlargement are also in keeping with what is typically found in the condition.[18,20] Atrial fibrillation (AF) which is present in up to one-third of patients was absent in the index patient.[19] There was also no evidence of AV block or ventricular arrythmias. The risk of sudden death in children with EMF is increased with the presence of arrythmias and our patient may be protected in this regard.[18,20] Holter ECG monitoring and frequent repeat electrocardiograms are however required to monitor for arrythmias.

Our patient was treated with diuretics and Angiotensin converting enzyme inhibitors which in addition to beta-blockers are the mainstay of medical treatment for this condition.[18-20] This treatment is largely symptomatic and of unproven benefit.[20] Medical treatment should act as a bridge to surgery. Anticoagulation or antiplatelet therapy is only recommended for patients with AF or for patients

with thrombi on echocardiography.[18] Endocardiectomy with valve repair is the surgical approach of choice and has proven effects on survival.[18,20] There is however a high operative mortality of up to 20% and open-heart surgery is currently beyond the reach of most patients in our setting.

CONCLUSION

The diagnosis of endomyocardial fibrosis has been made less frequently in the last two decades, even in areas endemic for the disease. It however remains an important cause of death due to acquired heart disease in children of the tropics and sub-tropics. The combination of late presentation, frequent misdiagnoses and poorly effective treatment options leads to an unacceptably high mortality. A high index of suspicion is critical.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Bassey K, Okpokowuruk FS, Oghenedoro O. Endomyocardial fibrosis in a 7-year-old Nigerian child: a case report and review of the literature. Int J Sci Rep 2022;8(6):151-4.