Isolated right ventricular endomyocardial fibrosis in a young male with chronic myeloid leukemia (RCD code: III-3F.2)

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Abstract

Endomyocardial fibrosis (EMF) is a rare disease of unknown etiology which is more prevalent in tropical countries. EMF is associated with fibrosis of endocardium, involving one or both ventricles. It usually presents with heart failure and has a poor prognosis. The diagnosis is usually made on echocardiogram, however cardiac magnetic resonance (CMR) is ideal for the diagnosis of EMF. We report a rare case of right ventricular endomyocardial fibrosis in a young male with chronic myeloid leukemia but no eosinophilia. Right ventricular EMF was diagnosed on the basis of classic findings on CMR. JRCD 2017; 3 (3): 98–100

Key words: rare disease, endomyocardial fibrosis, right ventricular failure, cardiomyopathy

Introduction

Endomyocardial fibrosis (EMF) is a type of restrictive cardiomyopathy of unknown etiology, usually found in tropical countries [1]. Davies and Conners first time described it in Uganda [2–4]. It is endemic in Sub-Saharan Africa, however cases have also been reported in Asia and South America [5]. EMF is characterized by fibrosis of the endocardium involving the apex and inflow part of both or one ventricle. Although the etiology of EMF is unknown, eosinophilia and immunopathologic processes probably play a role in its origin [6, 7]. Due to endomyocardial rigidity and progressive reduction in the cavity size of the affected ventricle, EMF results in heart failure.

Here we report a rare case of right ventricular endomyocardial fibrosis, from a non-tropical country.

Case Presentation

A 26 year old man was referred for cardiac magnetic resonance (CMR) for evaluation of right ventricular mass, found on multiple serial echocardiograms. He was known to have chronic myeloid leukemia (CML), BCR-ABL gene negative, for the past three years. There was history of gradually worsening dyspnea, bilateral leg swelling and abdominal distension for the last two years. He had been taking Imatinib and his primary disease i.e. CML was stable on medication. Clinical examination revealed mild bilateral pedal edema, ascites, hepatosplenomegaly and decreased breath sounds in the lower chest. Complete Blood Count showed hemoglobin of 13 g/dl, white blood cell count of 9760 cells/microliters (48% neutrophils, 39% lymphocytes and 5% eosinophils), consistent with the quiescent nature of CML. The patient had undergone multiple echocardiograms at other hospitals which revealed dilated right atrium and a right ventricular mass filling the major part of right ventricle including the apex. The right ventricular systolic function also seemed to be impaired due to the mass.

CMR with gadolinium enhancement was done at Siemens MAGNETOM Avanto 1.5 Tesla, which showed severely enlarged right atrium with some apical displacement of septal leaflet of the tricuspid valve and significant tricuspid regurgitation. Left ventricular volumes and systolic function were normal. Right ventricular volumes were: end diastolic volume (EDV) – 114 ml, end systolic volume (ESV) – 50 ml, stroke volume – 64 ml and right ventricular ejection fraction (EF) – 56%. There was obliteration of the right ventricular apex with a mass like density (Figure 1) which was isointense on turbo spin echo (TSE) T1 weighted images with and without fat suppression. It also appeared isointense on TSE T2 weighted images with mildly hyper-intense sub-endocardial
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rim. On first pass perfusion images after gadolinium injection, the obliterated apical region looked well perfused except a small superficial part which remained hypo-enhanced on early gadolinium images, suggestive of thrombus (Figure 2A & 2B). On delayed enhanced imaging with gadolinium (Figure 3A & 3B), a rim of endomyocardial hyper-enhancement was noted in the right ventricle that was consistent with endomyocardial fibrosis.

A diagnosis of isolated right ventricular endomyocardial fibrosis was made on the basis of classic findings on CMR. The patient was subsequently started on anticoagulation, small doses of furosemide, spironolactone and beta blockers in addition to Imatinib for his CML. Some improvement in clinical condition was observed after starting medical treatment.

**Discussion**

EMF is a type of restrictive cardiomyopathy. It is mainly a tropical disease; however cases have been reported in other regions of the world [5]. To our knowledge, this is the first case which is being reported from Pakistan.

The etiology of the disease is unknown, however it has been associated with a number of factors including tropical climate, helminthic infections and eosinophilia, certain cardiotropic viruses, ingestion of certain foods like cassava, combination of magnesium deficiency and high levels of cerium in the body [8–11]. No such factors were found in our patient. Although he was a case of CML but no eosinophilia was found and his CML was in a quiescent state on medications.

EMF is characterized by formation of fibrous tissue in the endocardium and to a lesser extent in the myocardium of one or both ventricles. The disease progresses in three stages, namely necrosis, thrombosis and fibrosis [12]. Clinically it presents as heart failure due to resultant restrictive cardiomyopathy. Our patient also presented with gradually worsening symptoms and signs of heart failure. The heart failure was predominantly right sided due to involvement of the right ventricle.

According to a study from Mozambique, biventricular endomyocardial fibrosis accounts for more than 55% of cases of EMF whereas 28% cases are those of right dominant EMF [13]. Isolated right ventricular disease is rare [14, 15] and a few case reports have been described in the literature. Patients with right dominant EMF usually present with pedal edema, ascites, cachexia and fatigue [16], as is the case in our patient.

In the previous case reports most cases of EMF were diagnosed on echocardiograms, which is a valuable tool for the evaluation of EMF [17]. Echocardiogram shows obliteration of the apex in
the affected ventricle along with thickened endocardium, sometimes resembling a mass. The right atrium is enlarged in right sided EMF along with tricuspid regurgitation. In the case presented here the right atrium was severely dilated along with tricuspid regurgitation and the echocardiogram reported right ventricular mass.

Recently, there has been a lot of interest in the use of cardiac magnetic resonance (CMR) in characterizing the features of EMF [18–20]. Late gadolinium enhancement using CMR delineates the endomyocardial fibrosis, which is the hallmark of EMF. CMR can also detect the presence of thrombus along with fibrosis. CMR was diagnostic in our case. This obviates the need for cardiac biopsy, which is not essential for the diagnosis of EMF [9].

EMF has a poor prognosis with no satisfactory medical or surgical treatment. Medical treatment includes low dose diuretics and anticoagulants. Our patient was also started on same medical treatment with some improvement in clinical condition. Surgical treatment for EMF includes resection of endocardial fibrotic tissue and valve repair or replacement [21]. We did not consider the surgical option as the surgical treatment usually acts as a palliative procedure and does not alter the disease [22].

References


Figure 3. Cardiac magnetic resonance. Delayed enhanced images with gadolinium, showing endo-myocardial hyper-enhancement in the right ventricular apex, indicated by the arrow