COEXISTENCE OF HYPERTROPHIC CARDIOMYOPATHY AND ENDOMYOCARDIAL FIBROSIS LEADING TO UNUSUAL BIVENTRICULAR HYPERTROPHY
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A 40 years old Afro-Caribbean woman from Nigeria living in Italy for a few months presented because of pelvic pain due to uterine myoma. In view of surgical treatment she had ECG, chest X-ray and standard blood test done. The ECG showed first degree atrio-ventricular block and pathologic Q waves in inferior leads. The radiograph documented some lung congestion. She was asymptomatic and the family history for cardiovascular disease was negative. Echocardiography was performed and revealed asymmetrical septal hypertrophy (maximal wall thickness 24mm) with some left ventricular (LV) apical hypertrophy and right ventricular (RV) involvement with apical obliteration. Differential diagnoses were considered, specifically hypertrophic cardiomyopathy (HCM) with biventricular hypertrophy, cardiac mass or tumor, endomyocardial fibrosis (EMF) with biventricular involvement or the coexistence of HCM with asymmetrical septal hypertrophy and EMF limited to the RV. Contrast echocardiography with Sonovue™ was employed to rule out RV hypertrophy at apical level. Contrast echo confirmed multiple fenestrations of the fibrous tissue over RV endocardium. Meanwhile we had the results of blood test and the blood count revealed a marked hypereosinophilia (17.5% - 1200/ml) restricting the possibilities to biventricular EMF and HCM associated with EMF. A cardiac magnetic resonance (CMR) with Gadolinium infusion was performed. The cine SSFP-CMR confirmed the asymmetrical septal hypertrophy with maximal wall thickness in the mid-posterior septum and RV obliteration. The tissue characterisation with gradient echo inversion recovery pulse after 10 minutes from Gadolinium infusion revealed two different scenarios. At the RV apex the LGE area in the endocardium and in the inflow tract as a continuous stria was typical for EMF. A cross cut in short axis with T2 weighted sequence showed hyperenhancement at the level of RV pavement suggesting oedema and then inflammation at this level. The cross cut at the same level with LGE sequence (fig. 2D) confirmed the fibrous tissue of the RV endocardium. Interestingly the cross cut at the level of the hypertrophied septum revealed subendocardial LGE involving the RV insertion point. This pattern of distribution (1) is common in sarcomeric hypertrophic cardiomyopathy (HCM) and suggests the coexistence of genetic cardiomyopathy. The patient underwent to RV endomyocardial biopsy that established the presence of significant fibrous subendocardial thickening with adherent thrombus containing sparse eosinophil cells suggesting thrombotic phase EMF. The genetic analysis revealed a known mutation of MYBPC3 gene (c.927-9 G>A) and confirmed the diagnosis of sarcomeric hypertrophic cardiomyopathy.

A single unifying diagnosis able to explain different aspects in a clinical scenario has regularly to be pursued by the clinical cardiologist but this is not always possible. In this case ECG and echocardiographic features did not support the diagnosis of biventricular HCM. CMR with Gadolinium helped to identify two different concomitant diseases through the capability to explore myocardial substrate and his role is well established in diagnosis and prognosis of EMF (2). However the gold standard in the diagnosis of EMF and HCM were endomyocardial biopsy and molecular biology respectively. HCM is the most common inherited cardiac disease with a prevalence of 1:500. In an endemic area of EMF as Nigeria, this association has to be presumed to be not rare even if to our knowledge this is the first case described in vivo (3).
References


Fig. 1: A) Long axis parasternal view: asymmetrical septal hypertrophy with maximal wall thickness 24 mm; B) Four chamber apical view: RV apical obliteration mimicking RV hypertrophy. Mild pericardial effusion and biatrial enlargement can be appreciated.
Fig. 2: A) SSFP sequence: the left ventricle presents asymmetrical septal hypertrophy with a global appearance consistent with hypertrophic cardiomyopathy. The apex of the right ventricle (RV) is obliterated and was suspicious for endomyocardial fibrosis; B) Gradient Echo inversion recovery sequence for Late Gadolinium Enhancement (LGE): the pavement of the RV shows hyper-enhancement suggestive of fibrosis. The arrows indicate the planes of short axis cross cut; C) T2 weighted sequence: cross cut in short axis at the level of RV pavement. The hyper-enhancement correlates with inflammation at this level and suggests active phase of endomyocardial fibrosis; D) LGE sequence: cross cut in short axis at the level of RV pavement. The hyper-enhancement in this sequence correlates with fibrosis; E) LGE sequence: cross cut at the level of maximal wall thickness of interventricular septum. LGE in subendocardium matching hypertrophied myocardium and involving RV insertion points is a pattern of distribution common in hypertrophic cardiomyopathy.
Fig. 3: A) Mallory trichrome stain: original magnification 200x - fibrous tissue in blue with poor cellular component and adherent thrombotic material on surface; 3B) Haematoxilin Eosin stain: original magnification 400x - in the context of thrombus, sparse eosinophil cells (arrow).