Hypertrophic Cardiomyopathy in a Middle Aged Man - A Case Report

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ABSTRACT

Background: Hypertrophic cardiomyopathy (HCM) is a disease of the myocardium with autosomal dominant pattern of inheritance. It is characterized by inappropriate hypertrophy involving either the interventricular septum, apex or left ventricular free wall in isolation or combined. The disease is often asymptomatic and may present for the first time with sudden death especially in young people. Recognition of the condition in our environment is often difficult because of dearth of facilities and expertise for echocardiography in most of our hospitals.

Objective: To draw attention to the existence of hypertrophic cardiomyopathy in our environment and need to use echocardiography in evaluating patients with cardiac diseases.

Method: The medical record of the patient and relevant literature were reviewed.

Result: A 47- year old civil servant presented for the first time with a 3- day history of severe, dull retrosternal chest pain and shortness of breath both of sudden onset associated with orthopnoea and paroxysmal nocturnal dyspnoea. He was managed for heart failure secondary to acute myocardial infarction based on clinical and electrocardiographic findings. He was unable to do echocardiography requested on initial presentation. Echocardiography was done 3 years later and revealed HCM. He however died shortly afterwards.

Conclusion: Though HCM is a relatively uncommon condition, it does occur in our environment and may mimic other disease conditions. The diagnosis requires a high index of suspicion, availability and utilization of echocardiography in investigating cardiac diseases.

Key words: hypertrophic cardiomyopathy, heart failure, myocardial infarction.

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INTRODUCTION

Hypertrophic cardiomyopathy (HCM) is a primary myocardial disorder with an autosomal dominant mode of inheritance characterized by abnormal myocardial hypertrophy. HCM is the most common inherited cardiovascular disorder and the commonest cause of sudden cardiac death in the young, especially young athletes. The overall prevalence of HCM is low and has been estimated to occur in 0.05-0.2% of the population.

Mutations in up to eleven genes that encode proteins of the sarcomeric apparatus of the myocardium have been implicated in HCM including beta and alpha myosin heavy chains, cardiac troponins and alpha tropomyosin. HCM may be asymptomatic well into adulthood and may present with sudden death at any age but commonly in young people. Other adverse presentation include heart failure, arrhythmias such as atrial fibrillation with consequent thromboembolism.

The clinical diagnosis of HCM is established most easily and reliably with 2- dimensional echocardiography. An unexplained maximum left ventricular wall thickness on 2- dimensional echocardiography greater than 15mm in any myocardial segment is sufficient to make diagnosis of HCM in adults. HCM is characterized by a small left ventricular cavity size (of <45 mm in end-diastole). The left ventricular cavity is enlarged only in the end-stage when it evolves into a hypokinetic, dilated and failing left ventricle.

CASE REPORT

Mr. OE, a 47-year old civil servant who was apparently in good health presented at the accident and emergency department of a tertiary hospital in Nnewi with history of sudden onset severe retrosternal chest pain which initially started at the back around the scapula and later migrated to the anterior chest and epigastric area. After a few hours he developed breathlessness associated with orthopnoea and paroxysmal nocturnal dyspnoea. He was not a known hypertensive or diabetic patient. He took alcohol and smoked cigarette occasionally. There was no family history of heart disease or sudden death.

On examination, he was in severe respiratory distress, afebrile, not pale and there was no pedal oedema. Cardiovascular examination revealed a pulse of 112 beats per minute small volume and regular. Blood pressure was 120/70mmHg. Jugular venous pulsation was not visibly elevated. Apex beat was located at the 6th left intercostal space at the anterior axillary line and not heaving. There was an S3 gallop. Respiratory rate was 40cycles/min. The chest had bibasal fine inspiratory rales. The abdominal examination revealed normal findings.

The investigations showed cardiomegaly on chest x ray. Electrocardiogram (ECG) showed sinus rhythm with rate of 100beats/min. Significant ST elevation was seen...
in lead II, III and AVF with pathological Q-waves. There was poor R wave progression in the precordial lead. Fasting blood sugar was 4.8mmol/L. Lipid profile, electrolyte, urea and creatinine were normal.

A diagnosis of acute left ventricular failure arising from acute myocardial infarction was made and he was treated with intravenous frusemide, aspirin 150mg, morphine, intranasal oxygen, lisinopril, digoxin, metoprol. His condition improved and he was discharged home after 19 days on admission. However, he was not regular with out patient clinic visit and was readmitted 10 months later with another episode of breathlessness. On examination, he was found to have irregular pulse. Electrocardiogram (ECG) revealed atrial fibrillation. He was managed as a case of congestive cardiac failure secondary to myocardial infarction with atrial fibrillation. He was treated with frusemide, spironolactone, digoxin, losartan and amiodarone. He improved clinically and was discharged after 39 days on admission. He subsequently defaulted on out-patient clinic visit and two years after the last admission he presented at the outpatient clinic with worsening breathlessness, orthopnoea, paroxysmal nocturnal dyspnoea and leg swelling. He was persuaded to do echocardiography which revealed grossly hypertrophic left ventricle with asymmetric septal hypertrophy. Inter-Ventricular Septum (IVS) was 24mm and Left Ventricular Posterior Wall (LVPW) was 14mm with IVS/LVPW of 1.7. There was Systolic Anterior Motion (SAM) of the anterior mitral valve leaflet. No regurgitation nor stenosis of the valves were noted. Ejection fraction was 58% with left ventricular diastolic dysfunction. The pericardium was normal.

Mr. OE was admitted for the third time from the outpatient clinic about three years after his first presentation. He however died 3 hours later.
DISCUSSION

Hypertrophic cardiomyopathy was initially thought to be a rare disease in black Africa in the pre-echocardiographic era. There are scanty reports on hypertrophic cardiomyopathy in Nigeria. The index case had symptoms commonly associated with hypertrophic cardiomyopathy such as chest pain and dyspnoea. This was in keeping with symptoms found in a study in Lagos by Mbakwem et al. over a 2-year period on patients referred for echocardiography, in which 14 cases met the criteria for HCM representing 1.9% of 714 subjects studied. About half of the patients had chest pain, palpitations and dyspnoea on exertion. However, 42.9% of the patients were asymptomatic. These symptoms found in HCM are non-specific and could be found in other cardiac diseases such as ischaemic heart disease. Mr. OE developed heart failure in the course of his illness. Heart failure has been reported to occur in HCM and is usually found in adults and results from diastolic dysfunction with a normal or supranormal left ventricular contractility. The case we presented had a normal ejection fraction of 58%, showing he had a normal cardiac systolic function. Paradoxically a small distinctive subset of HCM patients (5 - 10%) evolve into end stage (burnt out phase) characterized by left ventricular thinning, cavity enlargement and systolic dysfunction often resembling dilated cardiomyopathy and producing relentlessly progressive and irreversible heart failure.

The 12 lead electrocardiography is abnormal in 75% to 95% of patients with HCM and typically describes a wide array of patterns. We found S-T changes, pathological Q waves in the inferior lead, atrial fibrillation, atrial flutter, at different times in the case presented.

The treatment strategies in HCM include (a) medical therapy for mild to moderate symptoms, such as angina, heart failure, arrhythmias. (b) Surgical

FIG 2. M-mode echocardiography of the patient across the mitral valve showing Systolic Anterior Motion (SAM) of the anterior mitral valve leaflet. Arrows shows the leaflet making contact.
myectomy or myotomy (c) Alcohol septal ablation. (d) Implantable cardioverter-defibrillator’. Mr. OE was managed medically as he presented with heart failure. However, when the definitive diagnosis was made he did not live long enough for further treatment options to be explored.

In conclusion, this case is probably the first reported echocardiographically diagnosed HCM in our centre. The diagnosis was missed initially because he was unable to do echocardiography which was requested initially due to financial constraints. There is need for echocardiography to be readily available and affordable in order to make appropriate diagnosis of cardiac diseases and appropriate management instituted on time. The need for patients to keep their clinic appointment cannot be overemphasized to avert late presentation with high mortality.

REFERENCES