HYPERTROPHIC CARDIOMYOPATHY IN TWO YOUNG MEN: A CASE SERIES

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ABSTRACT

Background:

Hypertrophic cardiomyopathy (HCM) is a genetic disease of the myocardium with an autosomal dominant inheritance. The hallmark of HCM is left ventricular hypertrophy (LVH), which usually affects the interventricular septum in an asymmetric fashion; however, almost any pattern of LVH is possible including concentric LVH and hypertrophy localized to only one or two myocardial segments. The diagnosis requires a high index of suspicion because the disease is often asymptomatic and sudden death may be the initial presentation especially in young people.

Objective:

To highlight the importance of using echocardiography for evaluation of patients presenting with cardiovascular symptoms and the existence of hypertrophic cardiomyopathy in our environment.

Methods:

The medical record of the patients and relevant literature were reviewed.

Case Reports:

A 29 year old businessman and a 40 year old civil servant both presented with recurrent breathlessness on exertion, dull retrosternal chest pain, regular unprovoked palpitations and light headedness. Transthoracic echocardiography done on both patients revealed HCM.

Conclusion:

Hypertrophic cardiomyopathy is a cause of sudden cardiac death (SCD), though it is relatively uncommon, it does occur in our environment and may mimic other cardiac diseases.

Key words: Hypertrophic cardiomyopathy, left ventricular hypertrophy, sudden cardiac death.

INTRODUCTION

Hypertrophic cardiomyopathy is a genetic disorder of the myocardium caused by an autosomal dominant mutation in genes that encode sarcomere proteins or sarcomere associated proteins.[1] It is the most common genetic heart disease and evidence has shown that eight genes are known to definitively cause HCM: beta-myosin heavy chain, myosin-binding protein C, troponin I, troponin T, alpha tropomyosin, actin, regulatory light chain and essential light chain.[2,3] The overall prevalence of HCM is low and has been estimated to occur in 0.05-0.2% of the population,[4] with a mortality rate of approximately 1%.[5] HCM may be asymptomatic until adulthood. It is the most common cause of SCD in young people[6] and is responsible for 36% of sudden death in competitive athletes.[7]Most patients have obstruction of the Left ventricular outflow tract (LVOT) which may be present at rest or after physiological provocation in most patients with HCM.[8]

Electrocardiography (ECG) shows LVH in 75% to 95% of patients with HCM.[9] The clinical diagnosis of HCM is conventionally made with cardiac imaging, most commonly with 2-dimensional echocardiography (2D ECHO) and increasingly with cardiac magnetic resonance.[1] An unexplained maximal left ventricular wall thickness on 2D ECHO greater than 15mm in any myocardial segment is sufficient to make a diagnosis of HCM in adults.[10]

CASE SERIES

1.Mr OS, a 29-year old businessman who presented with a history of recurrent breathlessness on exertion of 2 years with a non-radiating retrosternal chest pain, regular unprovoked palpitations and light headedness. There was no syncope, orthopnoea, paroxysmal nocturnal dyspnoea or bilateral leg swelling. Past medical history revealed childhood hospital admissions for difficulty in

breathing and exemption from strenuous physical activities at school. He was not a known hypertensive or diabetic patient. He had significant alcohol and tobacco history. There was no family history of SCD or heart disease.

On examination, he was not pale, afebrile, not cyanosed and with no dependent oedema. Cardiovascular examination revealed a pulse of 72 beats/min, full volume and regular. Blood pressure was 120/80mmHg. Jugular venous pulsation was not visibly elevated. Apex beat was located at the 5^{th} left intercostal space, lateral to the mid-clavicular line and was heaving. There was an S4 with a grade 3 systolic murmur at the left sternal border and the apex. The respiratory rate was 18 cycles/min and the breath sounds were vesicular. The abdominal examination revealed normal findings.

The investigations showed cardiomegaly on chest radiograph. ECG showed sinus rhythm with significant ST elevation in the precordial leads from V1 to V3 with poor R wave progression, anterolateral and inferior walls myocardial ischemia. Cardiac Troponin I was elevated. Fasting lipid profile showed dyslipidaemia. Fasting plasma glucose was 4.2mmol/L. Uric acid, liver function test, electrolyte, urea and creatinine were normal. Echocardiography revealed grossly hypertrophied left ventricle with asymmetric septal hypertrophy (ASH). The interventricular septal wall thickness (IVS) was 30mm and the left ventricular posterior wall (LVPW) was 13.3mm with IVS/LVPW of 2.25 and a small left ventricular cavity (Figure 1 & 2). There was Systolic Anterior Motion (SAM) of the anterior mitral valve leaflet (Figure 3) and dilated left atrium. There was gross hypokinesia of the interventricular septum (IVS). Ejection fraction was 78.6% with restrictive left ventricular diastolic dysfunction. There was LVOT obstruction with a peak systolic pressure gradient between the left ventricle and the ascending aorta of 190 mmHg at rest. Medical treatment was started with a β blocker (tablets metoprolol 25mg twice daily), tablets rosuvastatin 10mg nocte and aspirin 75mg daily. Metoprolol was increased and maintained at 50mg twice daily. Other medications were continued at the same dose. He attended monthly follow up visits and made significant clinical improvement as his symptoms of palpitation, retrosternal chest pain and light headedness have resolved. He was then referred for implantable cardioverter defibrillator (ICD) for primary prevention of SCD.

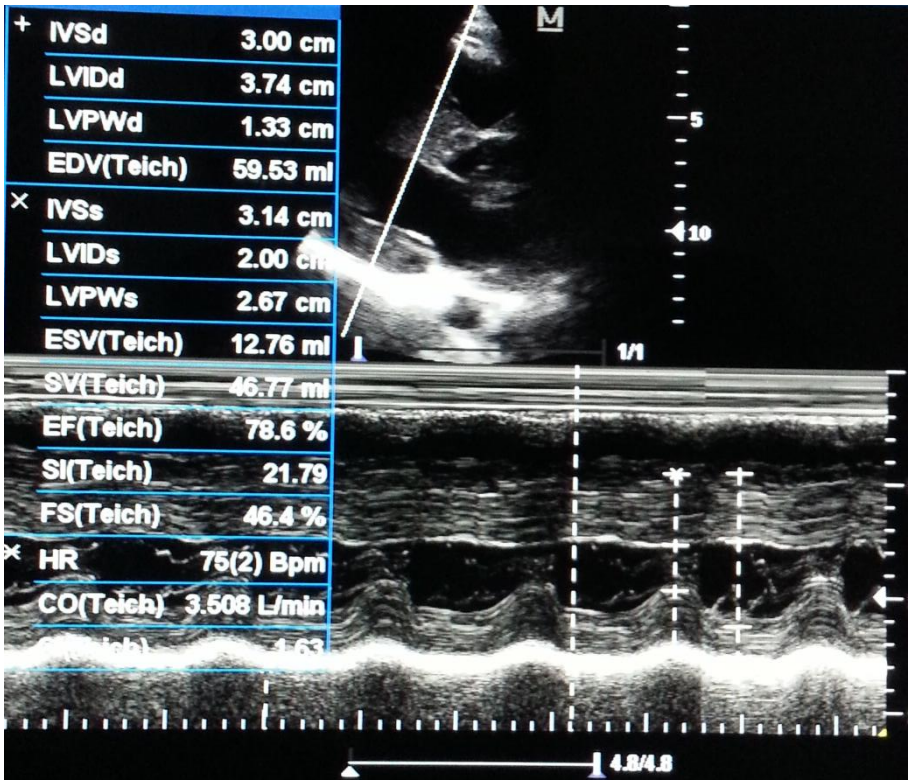

Figure 1: 2D guided M mode echocardiogram revealed grossly hypertrophied left ventricle with asymmetric septal hypertrophy (ASH) and small LV cavity.

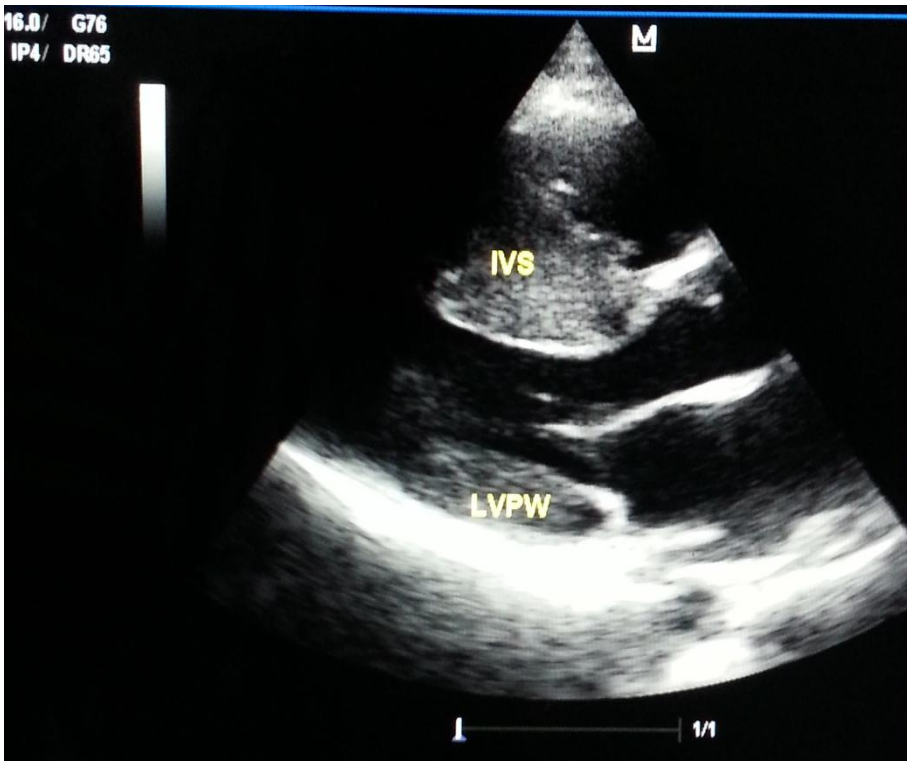

Figure 2: Parasternal long axis view showing markedly hypertrophied left ventricular septum in comparison to the posterior wall.

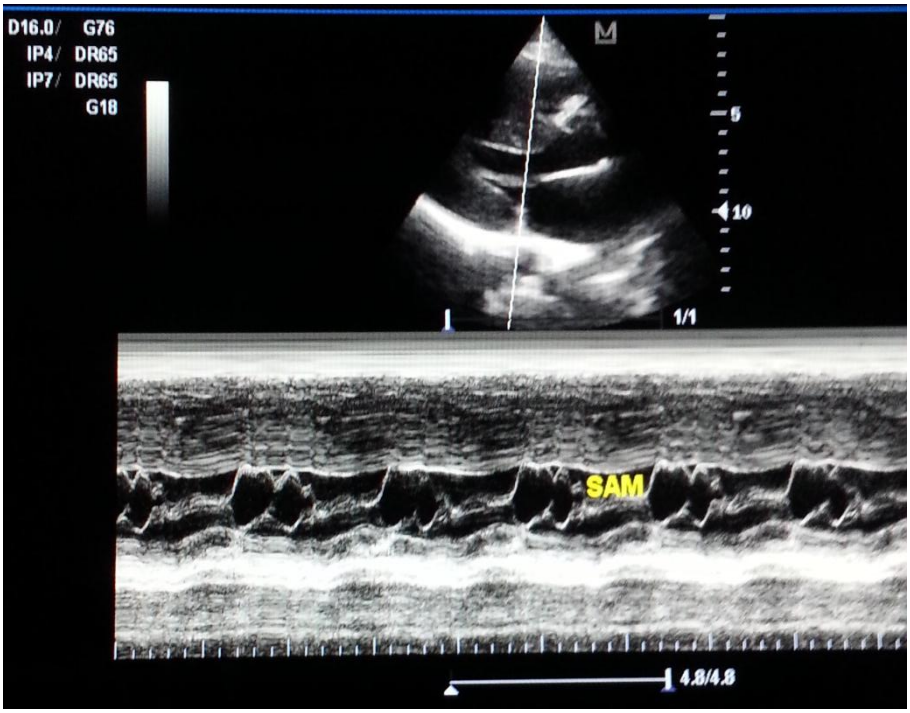

Figure 3: 2D guided M mode echocardiogram showing systolic anterior motion (SAM) of the mitral valve leaflet.

2. Mr AC a 40 year old civil servant who presented with a history of previous syncopal attack, recurrent light headedness, unprovoked palpitations, easy fatiguability and recurrent breathlessness. He was previously being evaluated for rheumatic heart disease. There was no orthopnea, paroxysmal nocturnal dyspnoea or bilateral leg swelling. He had a previous history of easy fatiguability from childhood and avoided strenuous activities since then. He was not a known hypertensive or diabetic. No significant alcohol or tobacco history. No family history of sudden cardiac death or heart disease.
On examination he was not pale, afebrile, not cyanosed, not dyspnoeic at rest with no dependent edema. His pulse was 92 beats per minute, full volume and regular. Blood pressure was 124/70mmHg. Jugular venous pressure was not elevated. The apex beat was located at the 5th left intercostal space lateral to the midclavicular line and was heaving. The 4th, 1st and 2nd heart sounds were heard. The breath sounds were vesicular with a respiratory rate of 16 cycles per minute. Electrocardiography showed ST depression and deep T wave

inversion in V4 – V6, AVL and I. It also showed left ventricular hypertrophy. Echocardiography revealed thickened interventricular septum of 2.27cm and left ventricular posterior wall of 2cm (Figure 4). There was systolic anterior motion of the anterior mitral valve leaflet (Figure 5) and dilated left atrium. The ejection fraction was 87.4%.The peak systolic pressure gradient between the left ventricle and the ascending aorta was 192mmHg. Laboratory investigations revealed dyslipidaemia but the fasting plasma glucose, uric acid and kidney function tests were normal. He was commenced on tablet rosuvastatin 10mg nocte, tablets aspirin 75mg daily and metoprolol 25mg twice daily. He was also counselled on the need for implantable cardioverter defibrillator but was lost to follow up.

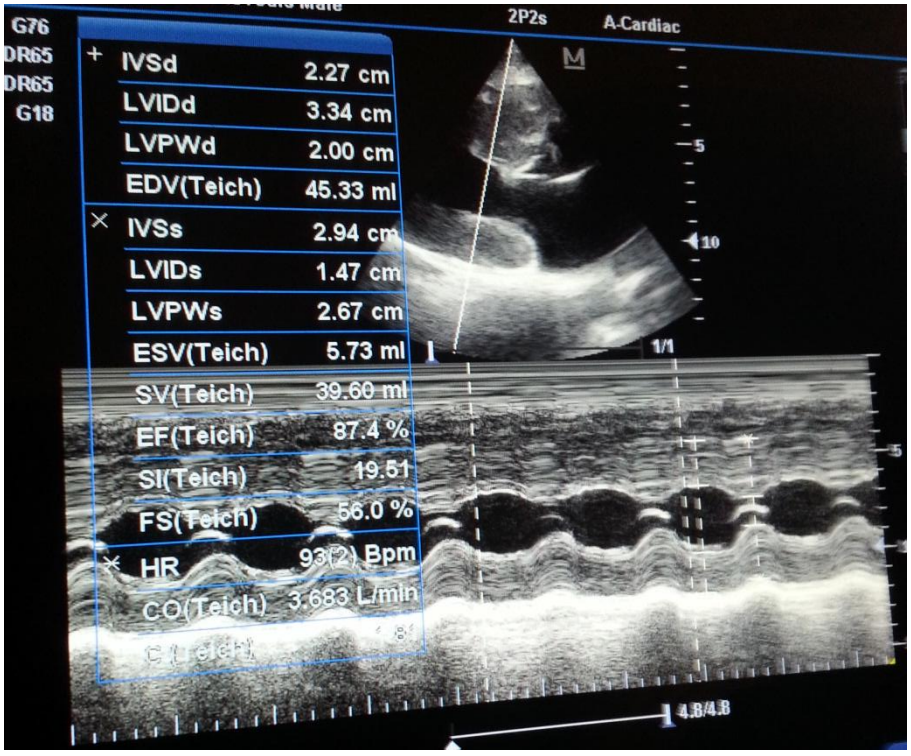

Figure 4: 2D guided M mode echocardiogram of Case 2 revealed thickened left ventricular walls.

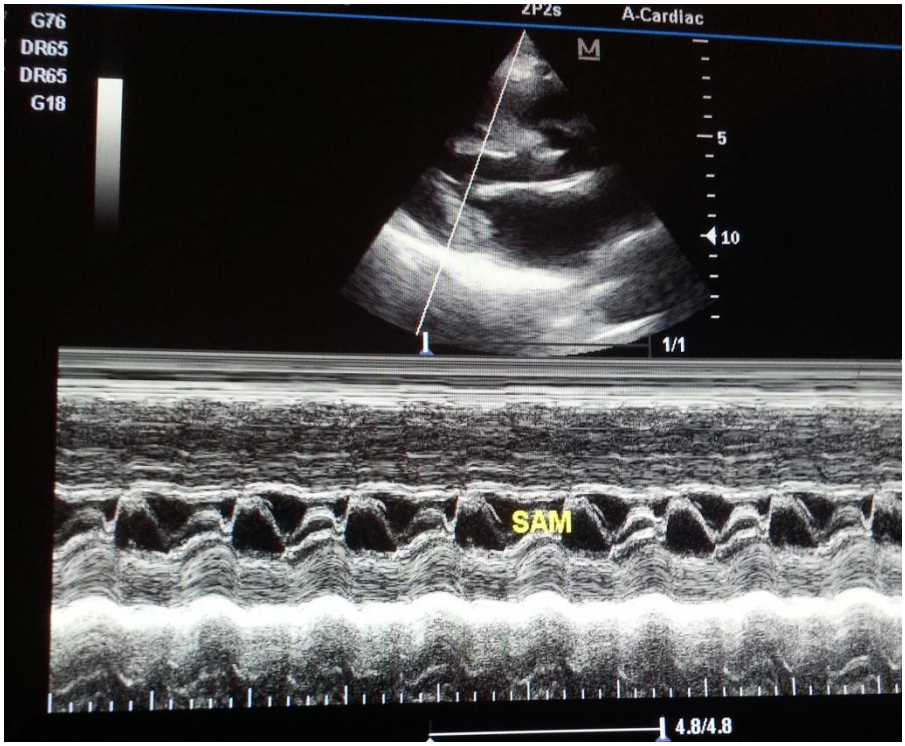

Figure 5: 2D guided M mode echocardiogram of Case 2 also showing SAM.

DISCUSSION

Hypertrophic cardiomyopathy was thought to be a rare disease in black Africans in the pre-echocardiography era[11] but this fact has been disputed by subsequent echocardiographic studies.[12,13] There is paucity of data on the prevalence of HCM in Nigeria, however Mbakwem et al[14] reported a much higher prevalence of 2% in Lagos. Both patients presented with dyspnoea on moderate exertion (DOE) in New York Heart association(NYHA) class ll. Dyspnoea on exertion, chest pain and palpitations are symptoms commonly associated with HCM. This was similar to the symptoms found by Mbakwem et al[14] in which half of the patients had palpitation and chest pain, while 42.9% had DOE and another 42.9% were asymptomatic. The symptoms found in HCM are non-specific and could mimic ischemic heart disease and other cardiac diseases.

The ECG and TTE in both patients were typical for HCM. The ECG for Mr OS showed significant ST elevation in the precordial leads from V1 to V3 which

was in keeping with an acute myocardial infarction. This was in keeping with the ECG found in a study in Germany by Daralammouri et al.[15] Other diagnostic modalities include a 48 hour ambulatory Holter ECG to detect non-sustained ventricular tachycardia and stress test.

Medical treatment with a beta adrenergic blockers or non-dihydropyridine calcium channel blocker which are known to decrease the obstructive gradient in HCM by decreasing cathecholamine –mediated contractility has been found useful in patients with mild to moderate symptoms[16]. Beta adrenergic receptor blockers are useful in relieving symptoms of heart failure in both obstructive and non-obstructive HCM by slowing the heart rate and reducing the force of left ventricular contraction thus augmenting LV filling and decreasing myocardial oxygen consumption.

Surgical septal myomectomy is done for symptomatic patients in New York heart association class lll /lV who do not respond to medical therapy[9]. It is also the indicated procedure in patients with obstruction to LV outflow under basal conditions or following physiological exercise.(LV outflow gradient ≥ 50mmHg)[1,5].During resection of part of the septal muscle and ventricular wall, complications with papillary muscle and left bundle branch block may occur[17].

For management of severely symptomatic patients with apical hypertrophic cardiomyopathy in this setting, Said and colleagues proposed a new surgical technique that they described as “apical ventriculotomy” to remove a portion of the thickened muscle to permit greater filling of the left ventricle in diastole[18]. When left ventricular outflow tract obstruction was present, an additional trans aortic resection was accomplished[18].

Percutaneous alcohol septal ablation has also been shown to reduce or eliminate the obstruction in 90% of cases.[9]

Dual chamber pacing has been promoted as an alternative to myomectomy for patients with refractory heart failure symptoms[5].

Implantable cardioverter defibrillator (ICD) is the mainstay of therapy for both primary and secondary prevention of SCD.[19] Sudden death is the most feared complication of HCM and may be prevented by the use of ICD. Secondary prevention is relevant in patients with a prior cardiac arrest and sustained ventricular tachycardia (VT). Indications for Primary prevention, especially in patients < 50yrs includes (a) A family history of 1 or more premature HCM related deaths particularly if sudden (b) hypotensive /attenuated blood

pressure response to exercise (c) unexplained syncope especially if recent (d) multiple or prolonged non sustained bursts of VT on serial or ambulatory ECG. (e)Massive LV hypertrophy ≥ 30mm. (f) Late gadolinium enhancement on cardiac magnetic resonance imaging.[6,19,20]

CONCLUSION

Hypertrophic cardiomyopathy does exist in our environment and may often be missed because of unavailability and poor utilization of echocardiography in the investigation of cardiac diseases. The diagnosis requires a high index of suspicion, therefore HCM should be considered in any young person presenting with unexplained chest pain, presyncope and dyspnoea on exertion.

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