

Variants of Hypertrophic Cardiomyopathy: A Registry Based Analysis of Twenty Cases

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ABSTRACT

Objective: The present research aimed at the careful evaluation of the different patterns, clinical presentation and establishment echocardiography as an authentic tool for diagnosis of Hypertrophic Cardiomyopathy (HCM) at an earlier stage.

Methods: In this cross sectional study 20 patients were enrolled after taking informed consent. Initially all patients had presented with atypical chest pain where 10 patients presented with exertional shortness of breath and 4 patients presented with acute breathlessness. Initial ECG revealed T inversion in anterior-inferior leads and upright T wave in aVR. Patients were initially diagnosed accordingly as Chronic stable angina, Heart failure with preserve Ejection fraction (HFpEF), hypertension and they underwent coronary angiogram (CAG) but CAG was normal in all patients. So, final evaluation of all patients done by repeat transthoracic echocardiography to find out the presence of any features of hypertrophic cardiomyopathy.

Results: Among 20 patients, males (85%) were more than females (15%) with mean age 56 ± 3 years. All 20 patients were diagnosed as chronic stable angina, 4 patients with heart failure and 12 patients with hypertension initially. Baseline T inversion in anterior-inferior leads and upright T wave in aVR. Echocardiography and coronary angiogram were normal in all 20 patients. Repeat echocardiography showed that about two-thirds (13) of them had apical HCM where one-fifth (4) had lateral wall hypertrophy and less than one-fifth (3) had septal HCM.

Conclusion: To minimize HCM related complications and earlier diagnosis, careful initial echocardiographic evaluation is an authentic tool.

Keywords: Hypertrophic cardiomyopathy; Echocardiography; Chest pain; Coronary angiogram

INTRODUCTION

Hypertrophic cardiomyopathy is a disease of myocardium where it becomes abnormally thick (hypertrophied) that makes harder to pump blood effectively. Very often it goes undiagnosed that can lead many people normal lives with no significant problems [1]. Although it is a common cause of sudden cardiac arrest in young people, including young athletes [2]. It is a genetic disorder where left ventricular hypertrophy remains unexplained

by secondary causes and a non-dilated left ventricle with preserved or increased ejection fraction [3]. It commonly affects people of any age with equal male-female distribution [4]. However, Mahony & Elliott claimed that there is male predominance (60%) in HCM with mentioning that women may be remained under-represented due to diagnostic bias with less hypertrophy and fewer electrocardiographic abnormalities. On the other hand, male predominance persists throughout the

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Received: July 16, 2020; **Accepted:** July 31, 2020; **Published:** August 7, 2020

Citation: Khaled MFI, Parvin T, Adhikari DK, Mahbub SME, Singha Ck, Khanal A, et al. (2020). Variants of Hypertrophic Cardiomyopathy: A registry Based Analysis of Twenty Cases. *J Clin Exp Cardiol* 11:666. DOI: 10.35248/2155-9800.20.11.666.

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range of left ventricular hypertrophy and is accentuated at the severe end of the spectrum [5]. Histologically, cardiac myocytes has pleiotropic nuclei with enlarged bizarre shapes. There is loss of normal parallel alignment because of haphazard orientation of hypertrophic myocytes that gives swirling appearance to the myocardial architecture with disarray. In HCM, disarray typically involves >10% of the myocardium with a predilection for the hypertrophied interventricular septum [4]. A small number of people with HCM presented with shortness of breath due to thickened myocardium, chest pain or problems in the heart's electrical system, resulting in lifethreatening abnormal heart rhythms (arrhythmias) [5]. Atrial fibrillation (AF) is the most common complication of HCM and is associated with an increased risk for morbidity and mortality. There are different types of hypertrophic cardiomyopathy: Anterior, lateral, septal and apical. Among them apical hypertrophic cardiomyopathy (ApHCM) is a rare form characterized by a spade-like left ventricular cavity. This rare form of hypertrophic cardiomyopathy more commonly involves the apex of the left ventricle and very uncommonly apex of the right ventricle or both [6]. Initially thought that ApHCM was unique among the Japanese population constitutes 15% of all HCM patients and about 3% among USA population but the incidence among the Asian population not insignificant [7]. The genetic basis of HCM is mutation of nine genes encoding for sarcomeric protein which shows the pattern of autosomal dominant inheritance in affected families [8]. Catastrophically HCM patients can present with sudden cardiac death (SCD) or ventricular arrhythmias or insidiously with symptoms of heart failure [3]. Patients with atypical chest pain, shortness of breath are often diagnosed as acute coronary syndrome (ACS) and treated with anti-ischaemic agents resulting no significant improvement. This misdiagnosis often leads to fatal outcome or sometimes delayed diagnosis of HCM was occurred. Echocardiographic features can be considered as an important diagnostic tool for HCM. So, the present research sought to the careful evaluation of the different patterns, clinical presentation and establishment echocardiography as an authentic tool for diagnosis of HCM at an earlier stage.

METHOD

This analytical research was conducted in the Department of Cardiology, University Cardiac Centre, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka from January 2019 to July 2019, after taking ethical clearance from the Institutional Review Board (IRB) of BSMMU, in accordance with Helsinki Declaration for Medical Research involving human subjects. An informed consent was taken from patients after informing details about the nature and purpose of the research.

Exclusion criteria

- Other concomitant systemic conditions that may lead to left ventricular hypertrophy.
- Patients with surgical or catheter septal myomectomy.

Operational definitions:

a. Hypertrophic Cardiomyopathy: ECG changes shows asymmetrical septal hypertrophy produces in the lateral (V 5-6, I, aVL) and inferior (II, III, aVF) leads. These may mimic prior myocardial infarction, although the Q-wave morphology is different: infarction Q waves are typically >40 ms duration while septal Q waves in HCM are <40 ms. Lateral Q waves are more common than inferior Q waves in HCM [2].

b. Apical Hypertrophic Cardiomyopathy: The diagnostic criteria for ApHCM included demonstration of asymmetric LV hypertrophy, confined predominantly to the LV apex, with an apical wall thickness ≥ 15 mm and a ratio of maximal apical to posterior wall thickness ≥ 1.5 mm, based on an echocardiogram or magnetic resonance imaging (MRI) [9].

After getting ethical clearance and consent, careful history and physical examinations were done in all 20 patients. Patients who were initially presents with atypical chest pain, shortness of breath or acute breathlessness enrolled in the research. All 20 patients had atypical chest pain where 10 with shortness of breath and 4 with acute breathlessness. Necessary investigations were done to evaluate the status of the patients. All the patients underwent electrocardiogram (ECG), echocardiogram and finally coronary angiogram (CAG). After necessary investigations, all patients were initially diagnosed as chronic stable angina (CSA). In addition to CSA, 4 patients were also diagnosed as heart failure (heart failure preserve ejection fraction- HFpEF) and 12 as hypertension (HTN). After evaluation all patients were proceeds with anti-ischaemic therapy. But the anti-ischaemic therapy had failed to improve the conditions. Finally researchers had decided to proceeds with ST segment changes in ECG. Repeat echocardiography was done to evaluate the patient's status. Careful evaluation of echocardiography was done to find out features of hypertrophic cardiomyopathy.

Statistical Analysis

Qualitative variables were expressed in percentages. Quantitative variables were expressed as mean \pm SD for normally distributed continuous variables. Analysis was done by MS Excel software

RESULTS

This analytical research was conducted in the University Cardiac Center, Bangabandhu Sheikh Mujib Medical University, Dhaka, over a period of one and half year from January 2015 to June 2016.

Table 1: Distribution of the sex among the patients

Sex	Frequency (n=17)	Percentage
Male	17	85
Female	3	15

n: Total number of patients

Data are presented as percentage (%)

Table 1 shows that more than two-thirds (85%) patients who participated in the research were male and rest of them were female with a mean age (SD. 56 ± 3 years) and the mean length of diagnosis from first medical contact was 5 ± 2 . All 20 patients were diagnosed as chronic stable angina, 4 patients with heart failure and 12 patients with hypertension.

Table 2 shows the ECG, echo and coronary angiogram findings that are observed in all 20 patients: All patients' presents with T wave inversion in anterior & inferior leads with upright T in aVR with normal findings in CAG. Echocardiogram shows the concentric left ventricular hypertrophy with no regional wall motion abnormality and good bi-ventricular systolic function.

Table 2: Investigations findings of patients

Name of the investigation	Frequency (n)	Major finding
ECG		T wave inversion
Echo	20	No wall motion abnormality
CAG		Normal

n: Total number of patients

Table 3 shows the echocardiographic findings of all patients. Among 20 patients, about two-thirds of them had apical HCM where one-fifth had lateral wall hypertrophy and less than one-fifth had septal HCM. Out of 20, increased left ventricular mid cavity gradient was found in 6 patients and maximum pressure was 135 mm Hg.

Table 3: Echocardiographic findings of patients (n= 20)

Echo finding	Frequency	Percentage (%)
Apical HCM	13	65
Lateral HCM	4	20
Septal HCM	3	15

n: Total number of patients

Data are presented as percentage (%)

DISCUSSION

The preset research aimed at to the careful evaluation of the different patterns, clinical presentation and echocardiography as an authentic tool for diagnosis of HCM at an earlier stage. Patients with atypical chest pain, shortness of breath or acute breathlessness initially diagnosed as CSA, HFpEF and HTN and treated accordingly but showed no significant improvement.

In this registry based analysis, male predominance was observed. HCM is one of the most common genetic disorders with a prevalence of one in 500 adults worldwide [10]. A research conducted by Velzen et.al showed similar findings with the

present research, male predominance up to 62% in all age group except in patients with more than 70 years where female predominance observed [11]. The mean age of all patients was (SD) 56 ± 3 which was due to the fact that age played an important determinant in the natural history of disease progression. Patients of advanced age group with HCM show an increase risk of heart failure, sudden cardiac death and consequences of atrial fibrillation [12]. Mean length of diagnosis from the first medical contact was more than 5 years, in this long period patients diagnosed as chronic stable angina and treated with antiischaemic agents without any significant improvement. It was fair enough to accept that longer duration of HCM without any confirmatory diagnosis may be due to the fact that sometimes ECG findings were normal initially with a gradual variable presentation. It was observed that about 6% of patient with normal ECG at earlier stage with a gradual appearance of left ventricular hypertrophy, ST and T wave abnormalities with pathological Q waves. Combined interpretation of echocardiographic findings and CMR imaging with age at diagnosis, inheritance pattern and associated clinical pattern can provide an initial clue for diagnosis of HCM at an earlier stage [13].

In ECG, all 20 patients presented with T wave inversion in anterior and inferior leads with upright T in aVR. Although ECG can make an essential tool for suspension of HCM but in 75% to 95% patients exhibit left ventricular hypertrophy and 25% exhibit a left anterior hemi block or left bundle branch block [14]. In the present research, about 65% patients were diagnosed as ApHCM. One of the most important findings for diagnosis of ApHCM was gaint T wave. Eriksson et. al) observed that 93% of the patients presents with negative T waves in precordial leads followed by 65% patients with left ventricular hypertrophy [9]. About 15% of the patients were diagnosed as septal HCM and 20% as lateral HCM. The incidence of sudden cardiac death and cardiovascular events are more common in patients with asymmetric septal hypertrophy than ApHCM [15].

CONCLUSION

To minimize HCM related complications and earlier diagnosis, initial echocardiography is an authentic tool. So, echocardiographers and clinical cardiologists should have sound knowledge regarding the echocardiographic findings related to hypertrophic cardiomyopathy (HCM).

LIMITATIONS OF THE PRESENT RESEARCH

1. Number of patients were few; more patients with same presentation will give more accurate results regarding different variants of HCM.
2. Single-centered analytical research.

CONFLICTS OF INTEREST

The authors have nothing to disclose.

ACKNOWLEDGEMENTS

The authors would like to thank all the patients who have participated in this cross-sectional observational research.

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