HOCM DIAGNOSTIC DILEMMA

Avinash Yashwant Pawar[1]

[1]*Senior Resident, Department of Cardiology, Dr. D. Y. Patil Medical College, Nerul, Navi Mumbai, Maharashtra.*

HOW TO CITE THIS ARTICLE: Pawar AY. HOCM diagnostic dilemma. J. Evid. Based Med. Healthc. 2018; 5(15), 1342-1344. DOI: 10.18410/jebmh/2018/278

PRESENTATION OF CASE

A 32 years old male, Type II diabetic patient was admitted in private nursing home in Mumbai in 2014 with h/o Chest Pain on exertion CCS class II, shortness of breath on exertion NYHA class II associated with intermittent palpitation since 2 months. Patient was comfortable at rest. There was no history of palpitations, syncope. His SpO_2 was 98%, pulse 90/min, BP 116/70 in right upper limb.

ECG showed Hyperacute T waves with ST elevation in all precordial leads. His CKMB, Tr T was in normal range. His Transthoracic 2D echocardiography showed mild concentric LVH, mild apical wall hypokinesia. Diagnosis of acute anterior wall myocardial infarction was made, and patient was thrombolysed with Reteplase. He was given Antiplatelets, statin and nitrates at the time of discharge. He was not prescribed beta blockers. Recently patient was evaluated at our center for similar symptoms. Clinically there was double apical impulse, systolic ejection murmur at neo aortic area. ECG had tall T waves with J point elevation. Echocardiography at our center showed HOCM with SAM and resting LVOT gradient of 30 mmHg. Diagnosis of HOCM confirmed by Cardiac MRI. Patients elder sister also died of cardiac illness at age of 38 years, but details of her illness not known to patient.

CLINICAL DIAGNOSIS

Hypertrophic Obstructive Cardiomyopathy which was misdiagnosed as AMI.

DIFFERENTIAL DIAGNOSIS

1. Acute myocardial infarction
2. Hypertrophic obstructive cardiomyopathy
3. Severe left ventricular hypertrophy

PATHOLOGICAL DISCUSSION

Hypertrophic cardiomyopathy (HCM) is a genetically determined heart muscle disease (60 to 70 percent) caused by mutation in one of the several sarcomere genes which encode components of the contractile apparatus of the heart. The prevalence of HCM in general population, as determined by echocardiographic studies around world is 0.2 percent (1 out of every 500 adults).[1,2] Hypertrophic cardiomyopathy is characterised by left ventricular hypertrophy of various morphologies, with various hemodynamic abnormalities like elevated filling pressure and impaired vasodilator reserve,[3] Depending on site and extent of cardiac hypertrophy it has 1. LV outflow obstruction 2. Diastolic dysfunction 3. Myocardial ischemia 4. Mitral regurgitation.

These structural and functional abnormalities can produce a variety of symptoms, including fatigue, dyspnoea, chest pain,[4] palpitations, presyncope or syncope. In broad terms, the symptoms related to HCM can be categorized as those related to heart failure (HF), chest pain, or arrhythmias. Patients with HCM has increased incidence of both supraventricular and ventricular arrhythmias and are at an increased risk of sudden cardiac death (SCD).

For the majority of patients with HCM, LVH is not progressive, and HCM is compatible with normal longevity in the west. Small group of patients however are at risk of sudden death, usually in the absence of symptoms. Progressive heart failure symptoms occasionally associated with systolic dysfunction; and atrial fibrillation with risk of thromboembolic stroke.

Histopathology in patients with HCM shows hypertrophied myocytes arranged in a chaotic and disorganised fashion with a varying amount of interstitial fibrosis. Intraluminal coronary arterioles are structurally abnormal with reduced cross-sectional area. There is also impaired vasodilatory capacity resulting in blunted myocardial blood flow during stress.

Newer techniques like genetic testing, cardiac magnetic resonance imaging,[5] have increased recognition of the HCM phenotype and improved clinical diagnosis. There is an autosomal dominant transmission of HCM which often involves multiple family members. Approximately 15 to 25 percent of patients with HCM report at least one episode of syncope. Another 20 percent complain of presyncope. Multiple mechanism may lead to an inadequate cardiac output or abnormal peripheral vascular reflexes.

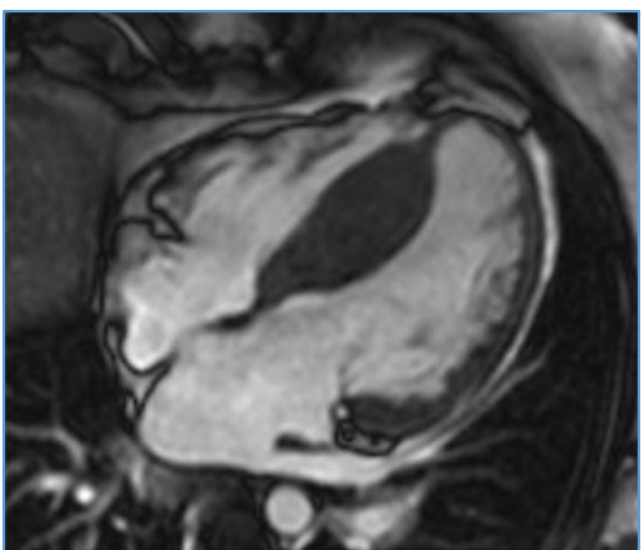
ECG should be performed in all patients with suspected diagnosis of HCM. ECG is the most sensitive routinely performed diagnostic test for HCM, but ECG abnormalities are not specific to HCM. A normal ECG is uncommon, seen in less than 10 percent of patients with HCM.[6] Typically, The ECG is abnormal with localised or widespread repolarisation changes. Prominent voltages with repolarization changes are typical of HCM associated with storage disease. Prominent abnormal Q waves, particularly in inferior (II, III, and AVF) and lateral (I, AVL, and V4-V6), P wave abnormalities, Left axis deviation, Deeply inverted T waves (giant negative T waves) seen in mid precordial leads (V2 –V4).[7]

Transthoracic echocardiography with two-dimensional, colour Doppler, spectral Doppler, and tissue Doppler should

Financial or Other, Competing Interest: None.
Submission 02-03-2018, Peer Review 10-03-2018,
Acceptance 06-04-2018, Published 09-04-2018.
Corresponding Author:
Dr. Avinash Yashwant Pawar,
#3C, 202, Royal Park CHS, Kanjur Marg,
East Mumbai-400042, Maharashtra.
E-mail: dravinashpawar@gmail.com
DOI: 10.18410/jebmh/2018/278

be done in all patients with suspected HCM. Transthoracic echocardiography can demonstrate cardiac morphology, systolic and diastolic function, presence and severity of left ventricular outflow tract gradient, and the degree of mitral regurgitation. Left ventricular hypertrophy is commonly seen in HCM. A clinical diagnosis of HCM is confirmed when unexplained LV wall thickness more than or equal to 15 mm is imaged anywhere in the left ventrical.[8] Systolic anterior motion of mitral valve. Sam of the mitral valve may result in LVOT obstruction when there is contact between the mitral valve and the septum. The grater the duration of mitral - eptal contact, the higher the LVOT obstruction. The presence of Sam is not required for diagnosis of HCM. LVOT obstruction is dynamic in nature, and is influence by factors which alter myocardial contractility and loading conditions (E.g.- dehydration, Ingestion of alcohol, heavy meals). Left atrium shows increased size, which can be associated with high risk for adverse disease related events in HCM, Including Atrial fibrillation. Stroke prophylaxis is required in such patients.[9] In majority of patients with HCM systolic function is normal, Often Hyperdynamic. Cardiopulmonary exercise testing can be done for assessment of peak VO_2. Increased left ventricular wall thickness in a pattern similar to that observed in sarcomeric HCM has also been observed in other diseases associated with mutation in genes related to carbohydrate metabolism, PRKAG2 and LAMP2. Although these disorders share a similar morphologic expression as patients with sarcomeric HCM, they also have some unique features and different natural history. Progressive conduction system disease requiring pacemaker implantation is common with PRKAG2 mutation, while progression to end stage heart failure and increased risk of ventricular tachycardia in early adulthood is common in males (X linked) with LAMP2 mutation. Approximately 25 percent of Noonan patients have increased left ventricular wall thickness similar to the pattern of hypertrophy observed in patients of sarcomeric hypertrophic cardiomyopathy, while Fabry disease may also mimic HCM. Screening for Fabry's disease among patients suspected HCM is required.
The major disease related complication of HCM ventricular arrhythmias leading to sudden death, chest pain, Progressive heart failure (HF) symptoms or HF death, Atrial arrhythmias including atrial fibrillation, and embolic stroke. In series published in 1980s the annual mortality of patients with HCM in referral center populations was 4 to 6 percent per year. However, lower annual mortality rates have been observed in most recent series from large unselected HCM patients population (approximately 1 percent or less per year) In a report from a referral population of 312 patients, 73 (23 percent) lived at atleast 75 years. HCM can now be considered a disease compatible with normal life expectancy for vast majority of patients with this disease. The annual rates of heart failure death or transplantation and stroke related deaths were 0.55 and 0.07 percent respectively. Published sudden death rates over the last 10 years of the study were lower than in previously published reports. The more recent studies were larger and included less severely affected patients as manifested by fewer patients with NYHA class III or IV heart failure and fewer patients who underwent septal myectomy. Similar outcomes have been reported in younger patients. Among a cohort of 474 patients younger than 30 years of age at presentation (mean age 20.2 years) who were evaluated at two referral centers between 1992 and 2013, the annual HCM related mortality rate was 0.54 percent per year over an average of 7.1 years of follow up. Additionally, 63 patients (13 percent of cohort; 1.8 percent per year) had aborted life- threating events (Including appropriate ICD interventions, resuscitated sudden cardiac arrest, or heart transplant)

In a series of 428 HCM patients presenting at age more than or equal to 60 years and followed for close to six years, risk was low for disease related morbidity and mortality, including sudden death (even with controversial risk factors). Non HCM related co-morbidities have greater impact on survival once hypertrophic cardiomyopathy achieves older age.



Four-chamber cardiac magnetic resonance (CMR) image showing massive septal hypertrophy in a patient with hypertrophic cardiomyopathy (HCM).

Figure 1

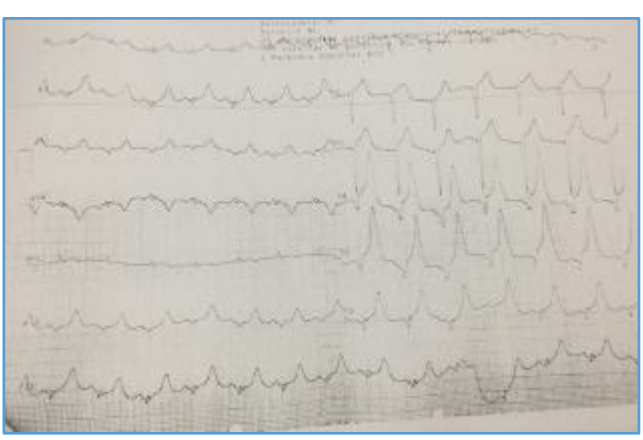


Figure 2

DISCUSSION OF MANAGEMENT

Medical management includes negative inotropic agents, including beta blockers, Nondihydropyridine calcium channel blockers (verapamil) and disopyramide.[10] Diuretics are

Relatively contraindicated in most patients with HCM due to potential reduction in preload, which may increase LVOT gradient, resulting in worsening symptoms and hypotension. However, in patients without LVOT obstruction who have refractory heart failure symptoms and are volume overloaded diuretics are effective in low dose. Ranolazine may be option for patients with ongoing symptoms inspite of medical therapy. Vasodilators such as dihydropyridine calcium channel blockers 9 eg, nifedipine, amlodipine), Nitroglycerin, Angiotensin converting enzyme inhibitors, angiotensin II receptor blocker, can produce fall in peripheral resistance with increased in LVOT obstruction and filling pressures, thereby resulting in hypotension and worsening of heart failure. Rate and rhythm control in tachyarrhythmias and atrial fibrillation Patients with HCM and atrial fibrillation have an increased risk of thromboembolism, regardless of CHA2DS2-VASc score, which is not applicable in this population as patients with HCM have been excluded from all trials of thromboembolism prophylaxis. Non-pharmacological treatment includes surgical myectomy and alcohol septal ablation. Up to 5 percent of patients with HCM will progress to end stage phase of the disease that is characterized by LV dilatation and thinning and systolic dysfunction. These patients are candidates for heart transplant along with optimum management for heart failure. For asymptomatic patients we suggest close clinical observation without any medical therapy.

HOCM may have diagnostic dilemma because of varied ECG presentation as we observed in this patient. There can be variable spectrum of presentation due to which this patient was misdiagnosed as Acute Myocardial Infarction and treated with thrombolytic agent.

But proper interrogation of symptoms on exertion, family history of sudden death of unknown cause with good clinical examination which had double apical impulse, ejection systolic murmur and Transthoracic echocardiography can clinch the diagnosis. Patient was given optimum dose of Beta blockers, advised to stop nitrates because it can reduce preload and increase LOVT gradient. Patient's counseling was done to avoid strenuous physical activity and about screening of other siblings. Increased left ventricular wall thickness in a pattern similar to that observed in sarcomeric HCM has also been observed in other diseases associated with mutation in genes related to carbohydrate metabolism, PRKAG2 and LAMP2. Although these disorders share a similar morphologic expression as patients with sarcomeric HCM, they also have some unique features and different natural history. Progressive conduction system disease requiring pacemaker implantation is common with PRKAG2 mutation, while progression to end stage heart failure and increased risk of ventricular tachycardia in early adulthood is common in males (X linked) with LAMP2 mutation. Approximately 25 percent of Noonan patients have increased left ventricular wall thickness similar to the pattern of hypertrophy observed in patients of sarcomeric hypertrophic cardiomyopathy, while Fabry disease may also mimic HCM. Screening for Fabry's disease among patients suspected HCM is required.

FINAL DIAGNOSIS

Hypertrophic Obstructive Cardiomyopathy

REFERENCES

[1] Maron BJ, Gardin JM, Flack JM, et al. Prevalence of hypertrophic cardiomyopathy in general population of young adults. Echocardiographic analysis of 4111 subjects in the CARDIA study. Coronary artery risk developments in (young) adults. Circulation 1995;929(4):785-789.

[2] Maron BJ, Mathenge R, Casey SA, et al. Clinical profile of hypertrophic cardiomyopathy identified de novo in rural communities. J AM Coll Cardiol 1999;33(6):1590-1595.

[3] Cannon RO, Rosing DR, Maron BJ, et al. Myocardial ischemia in patients with hypertrophic cardiomyopathy: contribution of inadequate vasodilator reserve and elevated ventricular filling pressures. Circulation 1985;71(2):234-243.

[4] Pasternac A, Noble J, Streulens Y, et al. Pathophysiology of chest pain in patients with cardiomyopathies and normal coronary arteries. Circulation 1982;65(4):778-789.

[5] Maron MS. Clinical utility of cardiovascular magnetic resonance in hypertrophic cardiomyopathy. J Carddiovas Magn Reson 2012;14:13.

[6] Veselka J, Anavekar NS, Charron P. Hypertrophic obstructive cardiomyopathy. Lancet 2017;389(10075):1253-1267.

[7] McLeod CJ, Ackerman MJ, Nishimura RA, et al. Outcome of patients with hypertrophic cardiomyopathy and a normal electrocardiogram. J Am Coll Cardiol 2009;54(3):229-233.

[8] Gersh BJ, Maron BJ, Bonow RO et al. 2011 ACCF/AHA guideline for the diagnosis and treatment of hypertrophic cardiomyopathy: executive summary: a report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines. Circulation 2011;124(24):2761-2796.

[9] Weissler-Snir A, Chan RH, Adler A, et al. Usefulness of 14-Day holter for detection of nonsustain ventricular tachycardia in patients with hypertrophic cardiomyopathy. Am J Cardiol 2016;118(8):1258-1263.

[10] Spirito P, Seidman CE, McKenna WJ, et al. Management of hypertrophic cardiomyopathy. N Engl J Med 1997;336(11):775-785.