Asymmetrical Septal Hypertrophic Cardiomyopathy: A Case Report

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Abstract

Background: Hypertrophic cardiomyopathy (HCM) is a clinically and morphologically heterogeneous genetic inherited cardiovascular disease that is often associated with unfavourable prognosis and leading to early death.
Case description: A young man suffering from chest pain with family history of sudden cardiac death . Electrocardiography showed left ventricular hypertrophy. Echocardiography demonstrated left ventricle concentric remodelling . However, exercise stress test and computed tomography-scan were unremarkable. Cardiovascular magnetic resonance imaging (CMR I) Eventually revealed the presence of l ate phase patchy myocardial enhancement at anteroseptal, septal, infero-septal in mid left ventricle , super io r and inferior left-right ventricle, asymmetrical insertion point of the left ventricle septum indicating HCM.
Conclusion: HCM is often found on young adults and mostly affected by genetic factor. Clinical examination and family history can assist the diagnosis of HCM. Echocardiography investigations can help direct HCM. CMRI is a more sensitive and less invasive diagnostic modality. Treatment includes beta blockers, septal myomectomy , and alcohol septal ablation. The latter is done frequently in Indonesia despite the high recurrence rate. Physical activity with light and moderate exercise can improve functional status and life quality in HCM patients.

Keyword: Hypertrophic, cardiomyopathy, asymmetrical, diagnosis, imaging.

1. Introduction

Hypertrophic cardiomyopathy (HCM) is an increased of left ventricular wall thickness and stiffness as well as changes in mitral valves and myocardial cells (Bos et al., 2009). Hypertrophic cardiomyopathy (HCM) is a genetically inherited disease of the heart muscle (60%-70%) caused by mutations in the sarcomere gene encoding for contractility (Bos et al., 2009; Dickstein et al., 2010). In the United States about 1 in 500 people have HCM in which increase the risk of sudden cardiac death especially in young people. HCM blocks the flow of blood from the heart to the rest of the body. Hence, the thickened heart muscle is too stiff to pump effectively (Dickstein et al., 2010) . Different types of HCM are classified by the part of the thickened heart wall as in apical hypertrophy, symmetrical hypertrophy, and asymmetrical septal hypertrophy with or without obstruction (Maron et al., 2003) . HCM is often found at a young age. In diagnosing HCM, a medical history from familial history and physical examination should be done. The Task Force for Diagnosis and Management of Hypertrophic Cardiomyopathy of European Society of Cardiology (ESC) supporting examinations can be done in electrocardiography (ECG), echocardiography, exercise ECG test, Holter monitoring, and cardiac magnetic resonance imaging (CMRI) (Dickstein et al., 2010; Maron et al., 2003)

2. Case Description

A 25-year-old man comes to the outpatient clinic of public hospital with a chief complain of chest pain that wakes him up at night. Over the last few months, the chest pain occurred during activities and rest. This complaint is accompanied by palpitations and dizziness. The patient has no complain of fainted, short of breath, nausea, vomiting, and cold sweat . In the past medical history, there was no history of hypertension, stroke , diabetes mellitus. In the familial history, patient's mother and maternal grandfather had sudden cardiac death. The patient occasionally drinks alcohol and smokes cigarettes.

On examination , the patient has palpitations with heart rates in 100 beats per minute while his blood pressure remains within normal limit at 110/70 mm Hg. His respiratory rates was 18 breaths per minute with oxygen saturation was 98% with free air . There was no jugular vein elevation. Cardiac examination showed regular rate and rhythm with single first and second heart sound without murmurs, gallops or extrasystoles . From pulmonary examination, auscultation both lungs was vesicular with no rhonchi or wheezing . There was no oedema in the upper and lower extremities. The electrocardiogram showed deep T wave inversion in the precordial leads and met the criteria for left ventricular hypertrophy . The chest radiograph shows a normal cardiac silhouette without pulmonary congestion. Figure 1 below depict the radiology and electrocardiography result.

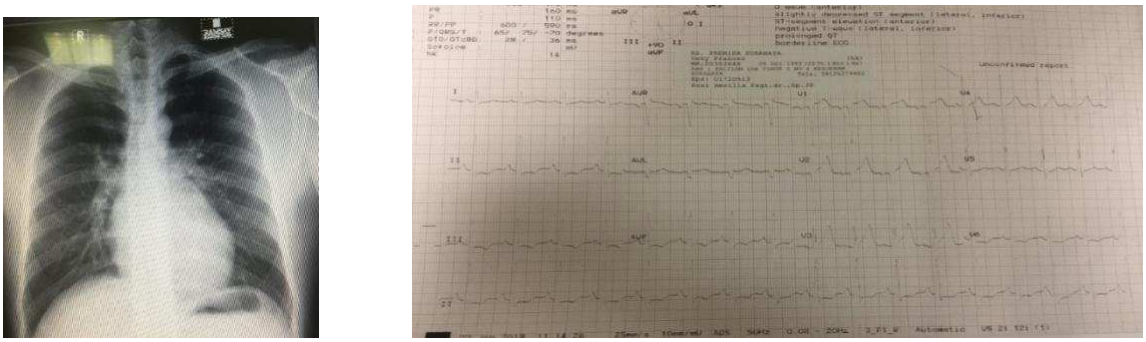

Figure 1. Chest X Ray and electrocardiography of the patient.

Cardiology consultation suggest to perform echocardiography. Transthoracic echocardiography showed no valve abnormalities with normal left ventricular ejection fraction of 70 % , LV concentric remodelling , diastolic dysfunction (abnormal relaxation) , septal hypertrophy, normal kinetic of LV segment and right ventricle function. Echocardiographic conclusion was normal LV volumes and ejection fraction with asymmetrical septal left ventricle hypertrophy (LVH) and LV concentric remodelling. This patient then underwent an exercise stress test resulting in normal stress test at 10.9 METs, no arrhythmias, negative ischemia response, and cardiorespiratory fitness. The patient also underwent a cardiac computed tomography scan resulting normal cardiac structure with no abnormalities were found.

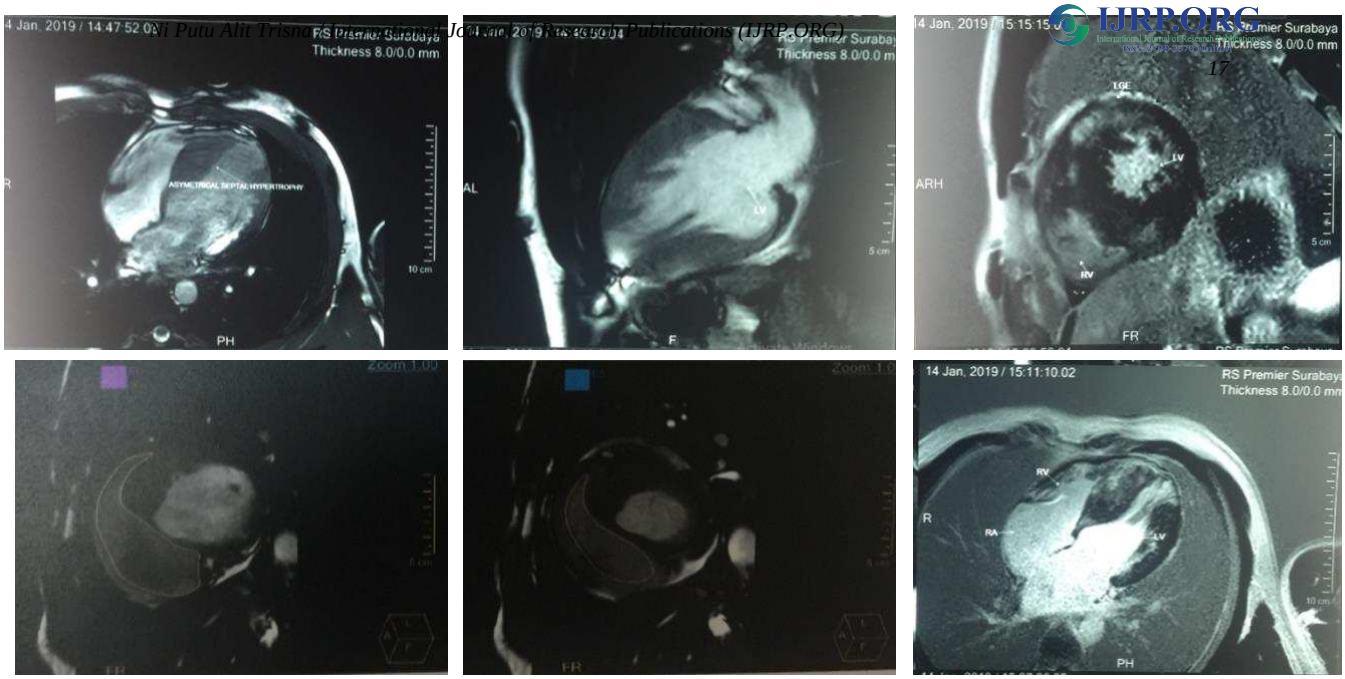

Figure 2. Cardiac magnetic resonance imaging results.

The patient was examined for viability with cardiovascular magnetic resonance imaging (CMR) which depicted in figure 2 resulting in left ventricle ejection fraction 62%, concentric LVH, diastolic dysfunction (abnormal relaxation), increased septal wall thickness (interventricular septum max 20.2 mm), increased LV mass , normal kinetic . Right ventricular ejection fraction normal 70%, normal kinetic. Normal atria and valves. Gadolinium study: early phase no thrombus in the ventricle . Late phase patchy myocardial enhancement at the anteroseptal, septal, inferoseptal mid LV, superior and inferior LV-RV insertion points. Pericardium normal shape and size, no effusion. Conclusion normal LV and EF volume, asymmetrical septal LVH, concentric LVH without systolic anterior motion (SAM) or LVOT obstruction, normal right ventricle ejection fraction , patchy myocardial fibrosis in anteroseptal, septal, inferoseptal . The patient was than temporarily treated with bisoprolol 1x 5 mg.

DISCUSSION

HCM was inherited by variated autosomal dominant. Cardiac troponin T, cardiac myosin binding protein C, and beta-myosin heavy chain are proteins that are most frequently involved in estimated 60% to 70% of all cases (Bos et al., 2009) . Additionally, in this case, there was a familial history of the mother who died suddenly at a young age. Histologically, there was myocardial disruption with cellular cardiomyocytes thickening up to 100 μm. The loss of contractility associated with impaired interstitial fibrosis which stimulate the process of myocardial hypertrophy. Relatively enlarged mitral valve leaflet to the left ventricular cavity is frequently developed.

HCM's pathophysiology consists of dynamic left ventricular outflow tract (LVOT) obstruction, myocardial ischemia, diastolic dysfunction, mitral regurgitation (MR), arrhythmias, and autonomic dysfunction. In some patients with certain HCM, clinical predominates (Fihn et al., 2014) . LVOT obstruction

occurred in more than 75% HCM involves septal hypertrophy with LVOT narrowing resulting in unusual blood flow vectors. A dynamic displacement of the anterior leaflet of mitral valve enhances the abnormal flow vectors. LVOT obstruction causes an increase in left ventricular systolic pressure which can magnify myocardial ischemia, LVH, and prolong ventricular relaxation. LVOT obstruction is linked to a reduced stroke volume and an escalated risk of heart failure. A more than 30 mm Hg peak LVOT gradient is indicating a significant obstruction. In patients with drug-refractory symptoms, a provoked obstruction of more than 50 mm Hg pressure gradient is the cut off indication for septal reduction therapy (SRT). The type of the obstruction should be characterised such as in dynamic LVOT obstruction, valvular, sub valvular, mid-cavitary gradient associated with papillary muscle hypertrophy, muscle obstruction caused by compensatory mid-ventricular hyperkinesis after apical infarction or papillary muscle attachment anomaly . Echocardiography examination is a standard procedure. Additionally, if the patient is suspected of having genetic HCM, echocardiography can be done every 12-18 months starting at the age of 12 years. Echocardiography should be done periodically when the patient is reaching puberty (Fihn et al., 2014) .

Altered ventricular load due to high intracavity pressure, abnormal intracellular calcium reuptake and imbalanced ventricular contraction and relaxation are common HCM disorders that cause diastolic dysfunction. Secondary MR from LVOT obstruction or from primary leaflet abnormalities often developed. SAM causes loss of leaflet coaptation. Hence, jets occur mostly in mid to late systolic posteriorly or laterally. This can help differentiate whether mitral valve SAM is the underlying cause of HCM and may be helpful if a septal myomectomy is performed (Fihn et al., 2014) . In this patient, SAM was not found from the TTE results.

HCM are susceptible to myocardial ischemia due to oxygen supply and demand imbalance . Hypertrophy of the medial and intramural arterioles, microvascular dysfunction with impaired coronary flow and myocardial hypertrophy can be seen in HCM patients (Geske et al., 2011). This abnormality is enhanced by hyperdynamic systolic function and LVOT obstruction with high intracavity pressure . This can be related to the prognosis of HCM patients with existing coronary atherosclerosis that can lead to infarction. In this patient, we can find concentric LV remodelling and LV asymmetry.

HCM patients may develop autonomic dysfunction, with compromised heart rate recovery and abnormal vasodilation. Abnormal blood pressure response to exercise described as a rise in systolic blood pressure failure by 20 mm Hg or decline in systolic blood pressure during exercise > 20 mm Hg from the optimal pressure was correlated with unfortunate outcome. Nevertheless, this inadequate response could be due to autonomy, abnormal diastolic filling, or LVOT obstruction. Abnormal blood pressure response can be attributed to surgical considerations which can be evaluated by a provocation test (Fihn et al., 2014) . In this patients the provocation test is normal.

Clinically, the presentation varies including syncope due to obstruction or tachyarrhythmias either ventricular or supraventricular, dyspnoea, dizziness or exercise intolerance. Chest pain can be resulted from myocardial and peripheral intramural coronary oxygen demand increase (Dickstein et al., 2010) . A systolic murmur can be detected by a variety of ECG findings including septal Q waves, negative precordial giant T waves, bundle branch block, and left ventricular hypertrophy,.

A high spatial and temporal resolution cardiac magnetic resonance imaging (CMRI) with concomitant administration of intravenous gadolinium contrast can non-invasively characterize myocardial tissue abnormality. In addition, CMRI excellently assess cardiac structural and functional capabilities. CMRI remains gold standard for assessing ventricular volumetric with additional value of evaluating myocardial fibrosis. Histological fibrosis and poor prognosis are pictured by hyperintense areas of the myocardium identified by late gadolinium enhancement (LGE). LGE is identified in up to 80% of HCM patients, usually in the interventricular septum or in the left ventricular wall at the right ventricular insertion points. LGE tends to be an independent risk factor for SCD in HCM. LGE is associated with the occurrence of ventricular tachycardia, suggesting that myocardial fibrosis is an arrhythmogenic substrate in HCM (Geske et al., 2011). We can find asymmetrical septal LVH in this patient.

Apical hypertrophic cardiomyopathy (APH) or localized myocardial hypertrophy (eg, lateral wall or inferior septum) that missed out by echocardiography are pinpointed by Steady-state free precession (SSFP). Cine SSFP demonstrate wall motion as well as jet turbulence across the LVOT in a patient with asymmetrical septal hypertrophy (ASH) HCM. These findings are linked to HCM-associated LVOT obstruction. Cine SSFP measuring myocardial mass and thickness accurately using retrospective gating (Geske et al., 2011). Cine SSFP sometimes shows a myocardium that is not hypertrophied or thin regions such as the basal crypt in HCM .

Basal asymmetrical septal hypertrophy as depicted in figure 6 is the most common cases of HCM. The anterior basal septal thickness is 15 mm at end-diastole and the septal to inferolateral ratio. In addition, MRI detect asymmetric hypertrophy with a spiral configuration: the anterior basal septum hypertrophies to apical inferior hypertrophy. LGE is usually identified with hypertrophy of the interventricular septum. Another type of ASH is a localized hypertrophy of the high septum or inferior septum is. High septal ASH is an indication for LVOT gradient reduction therapies such as alcohol septal ablation (ASA) and myectomy (Geske et al., 2011).

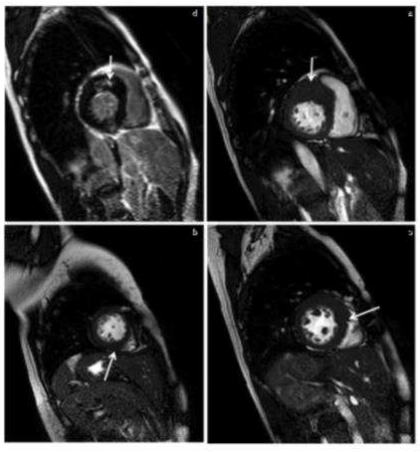

Figure 6. Short axis HCM asymmetrical septal

Midventricular obstruction (MVO) HCM as pictured in figure 7 has a bad prognosis due to considerable hypertrophy of the associated apical aneurysm and midventricular myocardium. Cine SSFP is also useful for distinguishing and midventricular hypertrophy MVO HCM . Further, LGE MRI shows the scar tissue replaced

apical aneurysm. Additionally, cerebral infarction can be caused by apical thrombus which associated with apical aneurysms in MVO HCM .

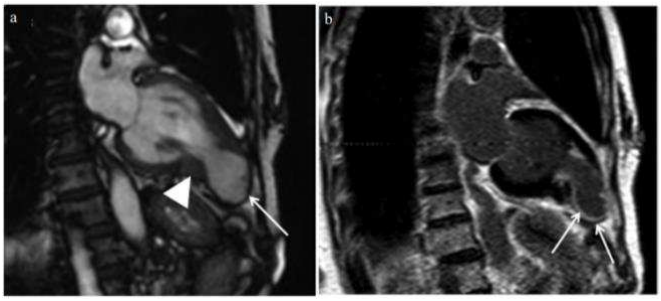

Figure 7. Four chamber midventricular obstruction in HCM

Local apical HCM of the apical myocardium and spade like deformity of the left ventricular cavity is shown in figure 8. It is known that APH shows T waves on the electrocardiogram. Cardiac MR imaging more frequently identifies this HCM phenotype than echocardiographic ones. Extensive LGE is associated with ventricular tachyarrhythmias even in cases of APH (Geske et al., 2011) .

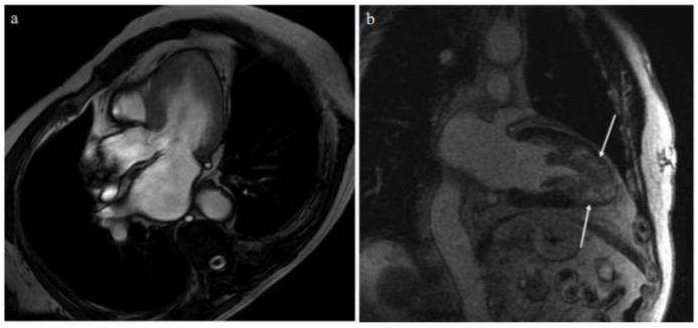

Figure 8. Apical in HCM

In one third of patients with HCM, there are right ventricular hypertrophy which concomitantly occur with left ventricular outflow obstruction. However, this type of HCM is unknown clinically.

Clinical Risk Factors for Risk Stratification of Sudden Death of HCM Family history of sudden death from HCM, massive LVH, unexplained syncope, HCM with LV systolic dysfunction, left ventricular apical aneurysm, extensive LGE on CMR imaging, NSVT on outpatient monitor. This patient had a family history of sudden death, LV systolic dysfunction and CMRI.

Pharmacological therapy of HCM according to the 2020 AHA with blockers (class IB), verapamil (class IB) was used if intolerance to blockers , calcium- channel blockers (class IIIC) and disopyramide (class IIaB). In atrial fibrillation conditions can be used digitalis (class IIIC) and the use of intravenous inotropes in hypotensive conditions (class IIIB) (Gersh et al., 2011; Bos et al., 2014; Elliott et al., 2014) . Invasive therapy can be in the form of septal reduction therapy (IC class) in the form of septal myomectomy and alcohol septal ablation (Geske, Ommen and Gersh, 2018).

Most patients with HCM can do activities or sports with light to moderate intensity to improve physical function, cardiorespiratory, and quality of life, as well as maintain health with physical activity guidelines. In patients with comorbid physician must prevent and treat comorbid to prevent severity of HCM such as obesity, atherosclerosis cardiovascular disease, impaired breathing during sleep and hypertension (Amano et al., 2018; January et al., 2019)

CONCLUSION

HCM often occurs at a young age, mostly due to genetic factors. Clinical examination and family history can help diagnose. Echocardiography may help to diagnose HCM. However, CMRI is better tool since it is more sensitive and less invasive. HCM therapy that is often used today can be medical or alcohol septal ablation. Light and moderate exercise can improve quality in HCM patients.

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