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Short Communication

Hypertrophic cardiomyopathy: in a search of its own identity

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ABSTRACT

Hypertrophic cardiomyopathy (HCM) is a genetic disorder characterized by left ventricular hypertrophy not explained by secondary causes, and a non-dilated left ventricle with preserved or increased ejection fraction. It is commonly asymmetric with most severe hypertrophy involving the basal interventricular septum. The histological features of HCM are myocyte hypertrophy and disarray, as well as interstitial fibrosis. The hypertrophy is associated with left ventricular diastolic dysfunction. Left ventricular outflow tract (LVOT) obstruction is present at rest in about one third of the patients, and can be provoked in another third. HCM is also an important cause of sudden cardiac death, particularly in adolescents and young adults. Non sustained ventricular tachycardia, syncope, a family history of sudden cardiac death, and severe cardiac hypertrophy are major risk factors for sudden cardiac death. Incidence being 30% and the causes of cardiac death are LVOT obstruction, increased oxygen demand of thick myocardium with compromised coronary circulation and intractable ventricular tachycardia. Mutations in over a dozen genes encoding sarcomere-associated proteins cause HCM. *MYH7* and *MYBPC3*, encoding β-myosin heavy chain and myosin binding protein C, respectively, are the two most common genes involved, together accounting for about 50% of the HCM families. Mutations in genes responsible for storage diseases cause a phenotype like HCM (genocopy or phenocopy). The routine applications of genetic testing and preclinical identification of family members represents an important advance. In genetic study HCM is associated with HLA DRW4.

Keywords: Hypertrophic cardiomyopathy, LVOT obstruction, Systolic Anterior Motion (SAM)

INTRODUCTION

HCM (Hypertrophic cardiomyopathy) is a asymmetrical septal hypertrophy where IVS thickness when compared to LV free wall is increased and the ratio between IVS and LV posterior wall is more than 1.33.It is an ECHO criteria by which it is being diagnosed[1]

It causes subvalvular aortic stenosis clinically. Apical hypertrophic cardiomyopathy is also a new entity, it was first time detected by Yamaguchi from Japan and there is disfigurement of the LV cavity. This may be associated with Systolic Anterior Motion (SAM) and without SAM on echocardiography[2]

HCM a genetic disorder of cardiac myocytes is characterized by cardiac hypertrophy, not explained by the loading conditions, a non-dilated left ventricle and a normal or increased ejection fraction. It is occasionally restricted to other myocardial regions, such as the apex, the mid-portion as well as the posterior wall of the left ventricle. At the cellular level, cardiac myocytes are hypertrophied, disorganized, and separated by areas of interstitial fibrosis. It is the relaxation of the ventricles which is affected, this is also known as superman's heart[3]

PATHOGENESIS

HCM is caused by mutations in sarcomere genes, diverse array of mechanisms, mirroring the diversity of the causal genes and mutations are implicated in the pathogenesis of HCM. The mechanistic events in HCM might be categorized into four sets of interlocking mechanisms. The primary defect is the mutation. Initial or proximal phenotypes are defined as those resulting from the direct effects of the mutations on the structure and function of the sarcomere proteins. Among the known causal genes, *MYH7* and *MYBPC* are the two most common followed by mutations in *TNNT2* and *TNNI3*. Rarely reported are mutations in genes encoding for other components of the sarcomere. The intermediary (or secondary) phenotypes include the molecular changes that occur in response to the changes in the sarcomere protein structure and function. Examples of the latter include altered gene expression and activation of the signalling pathways, such as the MAPK and TGFB1 pathways[4]

CLINICAL FINDINGS

It is an autosomal dominant conditions so all family members have to be screened as the family inheritance is 33%, it is more seen in males.

Chest pain, exertional dyspnea, syncopal attacks and sudden cardiac deaths are the usual presentations. On examination double apex beat is commonly seen, left parasternal heave of LV origin, patients pulse will be pulsus busferiencs which is 2 systolic beats, this is more commonly seen in patients with SAM which gives rise to subvalvular aortic outflow tract obstruction. Many times they are associated with ventricular arrthymias. Septal hypertrophy is more marked on the left ventricular side and sometimes RV compliance is also reduced when the hypertrophy spreads to the right side of the IVS. Patients will have 2 types of murmur because of left ventricular outflow tract obstruction giving rise to subvalvular aortic stenosis and the murmur is not associated with aortic systolic click. S4 sound can also be heard. Amyl nitrate inhalation will increase the intensity of murmur. Conduction of murmur in carotids is usually absent, murmur intensity is also increased in expiration due to increase in left ventricular stoke volume. Mumur of mitral regurgitation is also observed. There can be left atrial hypertrophy. Survival of more than 5 years after the onset of atrial fibrillation is very unusual. Due to the incomplete relaxation of the ventricle and reduced LV compliance there is a prominent diastolic dysfunction. Strenuous exercise can give rise to sudden death. Holter ambulatory monitoring becomes important in view of ventricular arrthymias[5]

ECG

ECG will show significant Q waves in septal leads: v1-v4 and in L2, L3, avf. Symmetrically inverted t wave is also an important feature. Bundle branch blocks are also seen in some of the cases, many of times they are associated with accessory pathway like WPW syndrome. Left atrial hypertrophy and left ventricular hypertrophy (voltage criteria) becomes important. LV original ventricular premature beats precedes before ventricular tachycardia, flutter and fibrillation. Medications which increase IVS contractility like sympathomimetic drugs or digitalis are to be avoided. Holter ambulatory will elicit ventricular arrthymias at rest and on exercise. Frequent runs of ventricular tachycardia and atrial fibrillation are also seen on holter[6]

ECHOCARDIOGRAPHY

On echo asymmetrical septal hypertrophy with reduced LV compliance and grade 1 to grade 4 diastolic dysfunction of LV will be seen. Systolic function of LV is always normal; the gradation of SAM decided the prognosis of the patient. Systolic anterior motion of the anterior mitral leaflet because of thick anterior papillary muscle signifies the outcome of the patient. The ratio of IVS and lVPD (Intra ventricular pressure difference) is more; 1.33 is the diagnostic criteria. Apical hypertrophic cardiomyopathy was detected first time from Japan by Yamaguchi where he has a study of 4 cases, here apical clots may be seen and they are usually asymptomatic, there the prognosis is not that bad and it may be an accidental study on 2D echo, LV angio shows reduced LV cavitatory size, color doppler gives gradation of mitral regurgitation. Coronary angiography except milking action will be normal in such patients. RV compliance may be reduced in some cases. All blood related family members should undergo 2D echocardiography. Another common finding is mild pericardial effusion-thin rim of anterior posterior echo free space in left parasternal long axis view is also an important and significant finding[7]

TREATMENT

Different classification of HCM like with obstruction and without obstruction of the left ventricular outflow tract, with SAM without SAM has been put forward, this entity was earlier classified into asymmetrical septal hypertrophy or Idiopathic hypertrophic sub aortic stenosis. Beta blocker: cardio selective and propranolol improves the LV function and decreased the LV outflow tract obstruction, similar encouraging reports have been put forward for diltiazem and verapamil (calcium antagonists) in life threatening ventricular arrthymias or severe left ventricular obstruction surgical treatment like septostomy or chemical ablation of septum by injecting alcohol into the septum with

permanent pacemaker implantation have shown encouraging results[8]

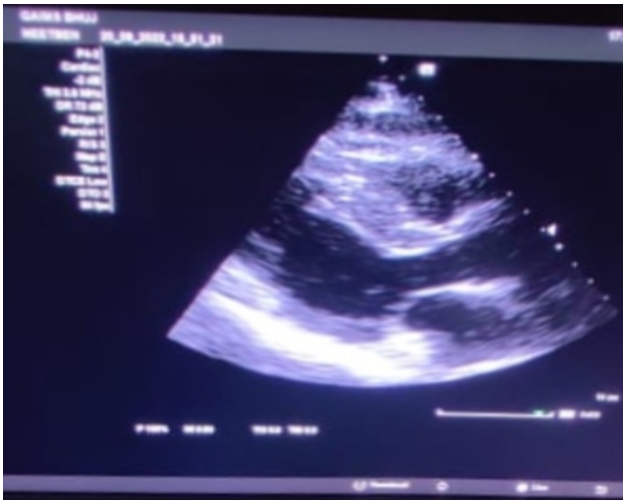

Figure-1: Parasternal long axis view showing decreased LV cavity, thick IVS and here the IVS to lVPW ratio is 1.6

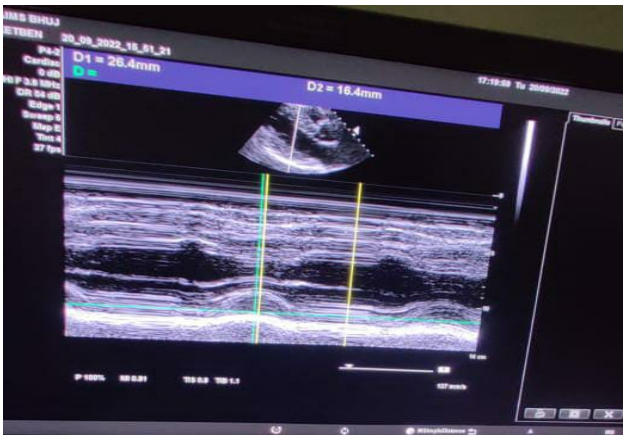

Figure-2: M-mode at the level of cordae tendinae showing ASH

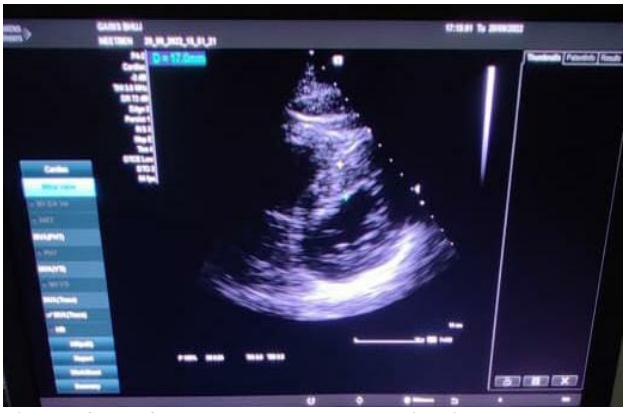

Figure-3: Left parasternal short axis view at the level of mitral valve

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