**APICAL HYPERTROPHIC CARDIOMYOPATHY WITH A RARE MYBPC3 GENE MUTATION VARIANT**

**P.T. Bhattacharya1**, T.A. Ahmad1, M.A. Bataineh2

1Mercy Catholic Medical Center, Drexel University, Philadelphia, PA, 2Drexel University, Philadelphia, PA, USA

Apical hypertrophic cardiomyopathy (HCM) is a unique form of HCM that is localized to the left ventricular apex with approximately 30% of patients found to be genotype positive that most often involve MYBPC3 and MYH7 genes. These patients are more likely to have family history for hypertrophic cardiomyopathy (HCM) but no additional HCM-related events are so far described. We are reporting a rare symptomatic patient with apical HCM who has MYBPC3 p.Gly596Arg mutation that was reported only once under the category of variant of unknown significance. A 49 year old male with no past medical history, presented with left sided chest pain. He has no family history of sudden cardiac death, syncope or cardiomyopathy. A 12-lead electrocardiogram revealed left ventricular (LV) hypertrophy and deep T wave inversions on anterolateral leads. Transthoracic echocardiogram showed a “spade-shaped” left ventricular apical hypertrophy, impaired diastolic relaxation, and preserved systolic function. Left heart catheterization showed elevated left ventricular end diastolic pressure of 20 mmHg and normal epicardial coronary arteries.LV angiogram showed apical hypertrophy with a spade-shaped left ventricle. Outpatient event monitor revealed 18 beat run of monomorphic non-sustained ventricular tachycardia and ventricular couplets. He had inducible polymorphic ventricular tachycardia by electrophysiology study and underwent implantation of an automatic implantable cardioverter defibrillator for the prevention of sudden cardiac death. Genetic testing revealed MYBPC3 p.Gly596Arg mutation.

Conclusion: Apical HCM is a unique variant of HCM where in reporting of genome-wide association studies and gene expression profiling are needed for better understanding of the genetic etiology of this disease.