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Beta-Myosin Heavy Chain (β-MHC) and Myosin Binding Protein C (MyBP-C) genes mutation in Bangladeshi hypertrophic Cardiomyopathy Patients: a genotype-phenotype correlation

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Abstract

Hypertrophic Cardiomyopathy (HCM) shows considerable clinical heterogeneity, both between and within families. So far 18 genes have been identified and about 1400 different mutations have been found. Among these mutations human beta-Myosin Heavy Chain (β-MHC) and Myosin Binding Protein C (MyBP-C) mutations account for about 50% of cases. In this study, Genotype and phenotype correlations in HCM in β-MHC and MyBP-C genes mutation were screened in 60 Bangladeshi HCM patients. Patients referred from Department of Cardiology, of BSMMU a tertiary care hospital, Dhaka, Bangladesh were included in this study. Clinical evaluation included a full clinical history, physical examination, 12-lead Electrocardiography, ambulatory holter monitor, and two dimensional M-mode echocardiography and Doppler echocardiography was done by expert cardiologist. Determination of genotype of β-MHC& MyBP-C genes were done by Miseq next gene sequencer (NGS). DNA was isolated from blood. Four primers for each gene were designed to cover the whole gene sequences and amplicons were prepared by long range PCR. Library preparation for NGS was done by Nextera XT library preparation kit. The barcoded libraries were sequenced using the MiSeq sequencer with v3 kits. All sequencing data were aligned automatically to the reference genome (GRCh37/hg19) using the MiSeq Reporter v2.5 which converted sequencing raw data to Binary Alignment/Map (BAM) and Variant Call Format (VCF) v4.1 files. The VCF files were comprehensively analyzed and interpreted by VariantStudio^{TM} v2.3 software. Evaluation of phenotype was completed after determination of genotype. Genetic screening revealed Glu965Lys/E965K, Arg442Cys/R442C (CGC>TGC), Arg663His/R663H (CGC>CAC) genes mutation in head domain of MYH7 protein in six patients, Ar435Trp/R35W (CGG>TGG) & Asp770Asn/D770N (GAC/AAC). Single gene mutations were identified in MYBPC3 protein in 15 patients and 3 patients found to be compound mutation. Two were found in intronic regions and are thought to be responsible for alternate splicing. Proband with compound mutations had a significantly greater left ventricular wall thickness and non-sustained ventricular tachycardia than the single mutation patients. Multiple gene mutation in HCM raises many issues of which haplotype of family member will be needed for microsatellites repeat to find out Bangladeshi population specific founder mutation.

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