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Association of MYBPC3 Promoter Polymorphism in Hypertrophic Cardiomyopathy

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ABSTRACT

Introduction:

Hypertrophic cardiomyopathy (HCM) is a disease of the myocardium characterized by thickening of the ventricles. The sarcomere is the structural and functional unit of cardiac muscle. SNPs/mutations in these disturb the contractile apparatus by altering the actin-myosin cross-bridge formation. The present study was aimed at screening for promoter SNPs in MYBPC3.

Materials and Methods:

The study includes 100 HCM patients and 100 controls for mutational screening by SSCP method and statistical & insilico analysis were performed to interpret the results.

Results:

Mutational Screening revealed 2 novel SNPs - **G22945A** and **A23211C of MYBPC3.** The **A allele of both polymorphisms** seem to confer **risk** towards HCM. This was further supported by insilico analysis which showed loss of transcription binding sites for 2 transcription factors, viz, Sox-5 and SRY in A23211C substitution of MYBPC3. Loss of transcription factor binding sites for SOX and SRY would result in delayed transcription and loss of several other transcription factors which bind to the SOX/SRY complex.

Conclusion:

The current study has shown 2 polymorphisms in the promoter region with detrimental effects towards the pathogenesis of HCM. It may be said that these polymorphisms may be linked to methylation patterns, and thus may act as regulatory mutations.

Key words: Hypertrophic cardiomyopathy, promoter, MYBPC3