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Sarcomeric Gene Variations and Phenotypic Plasticity of Cardiomyopathy

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ABSTRACT Hypertrophic Cardiomyopathy (HCM) is a primary cardiac disease characterized by increased thickness of the left ventricular walls in the absence of secondary causes and manifested by mutations in sarcomeric genes. Dilated Cardiomyopathy (DCM) is associated with dilation and impaired contraction of the ventricles and is associated with sarcomeric and cytoskeletal gene mutations. The present study is focused on screening for genetic variations in the sarcomeric genes viz., Myosin Heavy Chain gene (*MYH7*), Troponin I3 (*TNNI3*), Troponin T2 (*TNNT2*), Alpha-Actin (*ACTC*), Myosin Regulatory Light Chain (*MYL2*) and Myosin Essential Light Chain (*MYL3*) by SSCP and sequencing in 100 HCM, 97 DCM and 200 controls to elucidate the phenotypic plasticity associated with cardiomyopathy. Common variations were observed in exons 7, 12, 19 and 20 of MYH7; exon 9 and intron 16 of TNNT2; intron1, intron2, exon 5 and exon 7 of TNNI; exon 1 of MYL3 gene/s in both HCM and DCM cases. This can be explained on the basis of impaired energy compromise or dose effect of the mutant protein or environmental factors wherein a HCM could progress to a DCM phenotype affecting both the right and left ventricles, leading to heart failure. Genotype-phenotype correlations can be best explained by phenotypic plasticity where in mutations/ variations in the same gene and even same variations in a different environmental background may lead to HCM / DCM dispartite phenotypes.