ANGIOTENSIN-CONVERTING ENZYME INSERTION/DELETION POLYMORPHISMS CONTRIBUTES TO FAVORABLE CLINICAL COURSES OF HYPERTROPHIC CARDIOMYOPATHY CAUSED BY MYBPC3 GENE MUTATIONS

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Background: Hypertrophic cardiomyopathy (HCM) is a genetically heterogeneous disorder and differences in the clinical manifestations can be related to the presence of different disease-causing genes. MYBPC3 mutations carriers show favorable clinical courses compared to other disease-causing genes. Polymorphisms of angiotensin-converting enzyme insertion/deletion (ACE I/D) is associated with left ventricular (LV) hypertrophy and remodeling, and could influence the difference in clinical courses of genotyped HCM.

Methods: We studied 126 HCM mutation carriers of 4 disease-causing sarcomere gene namely MYH7 (n=17), MYBPC3 (n=35), TNNT2 (n=23), and TNNI3 (n=51) including typical and end-stage HCM (64 males, mean age 51±21 years). Relationships among underlying sarcomere gene, ACE I/D and echocardiographic parameters were examined.

Results: MYBPC3 mutation carriers showed highest ejection fraction (EF), smallest LV dimension and smallest proportion of end-stage among 4 disease-causing genes, but not significant. ACE II genotype exhibited significantly smaller LV end-systolic dimension (28±7mm vs 32±11mm, p<0.05), higher EF (62±12% vs 56±15%, p<0.05) and smaller proportion of end-stage (9% vs 26%, p<0.05) than those of D allele. Frequency of II genotype was higher than that of D allele only in MYBPC3 mutations carriers (p<0.05).

Conclusion: Frequency of ACE II genotype, which showed benign phenotype, was higher than that of D allele only in MYBPC3. This may contribute to favorable clinical courses of MYBPC3 mutation carriers compared to other disease-causing gene.