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Significance of sarcomere gene mutation in patients with dilated cardiomyopathy

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ABSTRACT. Dilated cardiomyopathy (DCM) is a myocardial disease with a high mortality rate. Approximately 40 genes have been found to be associated with DCM to date. Non-familial DCM can also be caused by gene mutations, suggesting that genetic factors were involved in the pathogenesis of DCM; therefore genetic testing is beneficial for the early diagnosis of DCM, which can facilitate the implementation of preventive measures by and within patient's families. Here, we investigated the underlying genetic mutations involved in the cause of patients with DCM. This prospective study included 240 patients with idiopathic DCM and 240 healthy volunteers. Subject clinical data were collected and polymerase chain reaction amplification was carried out on subject DNA for three candidate genes tropomyosin (*TPM1*), cardiac troponin T type-2 (*TNNT2*), and nuclear lamina protein A/C.

Single nucleotide polymorphism (SNP) loci were detected in the *TPM1* (rs1071646) and *TNNT2* (rs3729547) genes, respectively. The genotype distributions and allele frequencies were found to satisfy Hardy-Weinberg equilibrium, which indicated that the group was representative. Statistically significant differences were found between the variant frequencies in the two SNP loci between the Kazakh patients with idiopathic DCM (IDCM) and healthy volunteers. A significant difference in the genotype distributions (P = 0.000) and allele frequencies (P = 0.000) of SNP rs1071646, and another significant difference in the genotype distributions (P = 0.000) and allele frequencies (P = 0.039) of SNP rs3729547 between Kazakhs with IDCM and Kazakh controls. These results suggest that the *TPM1* (rs1071646) and *TNNT2* (rs3729547) gene variants might represent risk factors for patients with DCM in the Kazakh population.

Key words: Dilated cardiomyopathy; Cardiac troponin T type-2 gene; Gene mutation; Single nucleotide polymorphism; Tropomyosin gene; Nuclear lamina protein gene