Association between -44G/A and +71A/G polymorphisms in the connexin 40 gene and atrial fibrillation in Uyghur and Han populations in Xinjiang, China


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ABSTRACT. We aimed to elucidate the association between connexin 40 (Cx40) genetic polymorphisms and atrial fibrillation (AF) in a Chinese population in Xinjiang comprising Uyghur and Han individuals. We enrolled 275 Uyghur and 305 age- and gender-matched Han subjects, and used polymerase chain reaction to detect single nucleotide polymorphisms (SNPs; -44G/A and +71A/G) in the gene encoding Cx40. A mutation screening was performed by direct sequencing and calculation of genotype and allele frequencies among AF patients and control subjects to determine the relationship between these variants and this condition in Uyghur and Han populations. The two SNPs examined were significantly associated with AF in both
ethnic groups. Further analysis showed the SNPs to be in perfect linkage disequilibrium in both AF and control groups among Uyghur and Han individuals. In both populations -44AA genotype and A allele frequencies among AF patients were significantly higher than those in the control group. In addition, under the dominant model (GG vs GA+AA), a significant difference in the distribution of Cx40 -44G/A genotypes was detected between patients and controls. Logistic regression analysis revealed that Cx40 genetic polymorphisms increase AF risk in Uyghur and Han residents of Xinjiang. In conclusion, both the -44G/A and +71A/G variants of the gene encoding this protein are associated with AF in Uyghur and Han populations in northern China.

**Key words:** Atrial fibrillation; Connexin 40; Gene polymorphism