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A NOVEL POLYMORPHISMS IN THE ABCA1 GENE M233V IN MONGOLIA POPULATION

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Objectives To investigate whether ATP-binding cassette transporter 1 (ABCA1) M233V genetic variation is correlated with blood lipids in Mongolian population and the association of this polymorphism with coronary heart disease (CHD).

Methods The target fragments of ABCA1gene was amplified and analysed by PCR-restriction fragments length polymorphism (RFLP) technique in 115 Mongolian control subjects without CHD and patients with CHD.

Results A novel polymorphism in the ABCA1 gene Was found in 32 patients: M233V which exists in exon7 of ABCA1 gene and it's cDNA location is A1092G and converses 233 amino acid from Methionine to VMefie. ABCA1 gene M233V polymorphism were existing in Mongolian population, which had three types MM genotype, MV genotype and VV genotype. The highest frequency of ABCA1 M233V genetic variation was MM genotype, the next was MV genotype and the lowest was VV genotype in Mongolian population. There was no significant difference in frequency of allele and genotype in M233V polymorphism between controls and CHD patients (p>0.05). No significant difference was found in level of TC, TG, HDL-C, LDL-C between MM genotype and MV+VV genotype (p>0.05).

Conclusions M233V is a novel polymorphism in the ATP-binding cassette transporter A1 gene. The highest frequency of ABCA1 M233V genetic variation was MM genotype, the next was MV genotype and the lowest was VV genotype in Mongolian population.

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