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RELATIONSHIP BETWEEN A NOVEL POLYMORPHISM OF C5L2 GENE AND CORONARY ARTERY DISEASE IN CHINESE HAN AND UYGHUR POPULATION

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Background C5L2 was demonstrated a functional receptor of acylation-stimul- ating protein (ASP) which is a stimulator of either triglyceride synthesis or glucose transport. However, little is known about the variations in the coding region of the C5L2 gene and theirs association with coronary artery disease (CAD). **Methodology/principal findings** In the present study, we identified a novel single nucleotide polymorphism (SNP), 698 C>T (P233L), in exon 2 using PCR-directly sequencing method. This nucleotide change causes the amino acid change from proline to leucine at codon 233. We examined the role of this SNP for CAD using a 2-stage design, first we determined the prevalence of C5L2 genotypes in 492 CAD patients and 577 age

and sex-matched healthy control subjects of Han population, Second, a replication analysis of 319 CAD patients and 554 age-and sex-matched healthy control subjects of Uyghur population. Heterozygote carriers of the 698CT genotype were more frequent among CAD patients than among controls in Han (7.3% vs 1.7%) and in Uyghur population (4.7% vs 1.6%), respectively. The OR for carriers of this genotype for CAD was 4.484 (95% CI 2.197 to 9.174) in Han and 2.989 (95% CI 1.292 to 6.909) in Uyghur population, respectively. After adjustment of the confound factors such as sex, age, smoking, drinking, hypertension, diabetes, triglyceride, total cholesterol, high density lipoprotein, the difference remains significant both in Han (p<0.001, OR 6.604, 95% CI 2.776 to 15.711) and in Uyghur population (p=0.047, OR 2.602, 95% CI 1.015 to 6.671), respectively.

Conclusion/significance We conclude that the 698CT genotype of C5L2 may be a genetic maker of CAD in Chinese Han and Uyghur population.