RELATIONSHIP BETWEEN A NOVEL POLYMORPHISM OF THE C5L2 GENE AND CORONARY ARTERY DISEASE

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Background C5L2 has been demonstrated to be a functional receptor of acylation-stimulating protein (ASP), which is a stimulator of triglyceride synthesis or glucose transport. However, little is known about the variations in the coding region of the C5L2 gene and their association with coronary artery disease (CAD).

Methodology/Principal findings The authors identified a novel single nucleotide polymorphism (SNP), 698C>T (P233L), in exon 2 using a PCR direct-sequencing method. This nucleotide change causes the amino-acid order from proline to leucine at codon 233. We examined the role of this SNP for CAD using two independent case–control studies: one was in the Han population (492 CAD patients and 577 control subjects) and the other was in the Uygur population (319 CAD patients and 554 control subjects). Heterozygote carriers of the 698CT genotype were more frequent among CAD patients than among controls in the Han population (7.3% vs 1.7%) and in the Uygur population (4.7% vs 1.6%). The odds ratio (OR) for carriers of the 698CT genotype for CAD was 4.484 (95% CI: 2.197 to 9.174) in the Han group and 2.989 (95% CI: 1.292 to 6.909) in the Uygur population. After adjustment of confounding factors such as sex, age, smoking, alcohol consumption, hypertension, diabetes, as well as serum levels of triglyceride, total cholesterol, high-density lipoprotein, the difference remained significant in the Han group (p<0.001, OR=6.604, 95% CI: 2.776 to 15.711) and in the Uygur group (p=0.047, OR=2.602, 95% CI: 1.015 to 6.671).

Conclusion/Significance The 698CT genotype of C5L2 may be a genetic marker of CAD in the Han and Uygur population in western China.