

胆固醇7 α -羟化酶基因A-204C单核苷酸多态性及其与血浆血脂的关联 Association of 7 α -hydroxylase gene polymorphism with levels of plasma lipids

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摘要

为探讨胆固醇7 α -羟化酶基因(CYP7A1)多态性与中国四川汉族人群的分布及其与冠状动脉粥样硬化性心脏病(coronary heart disease, CHD)易感性的关系, 对183例CHD患者和101例对照用聚合酶链反应-限制性片段长度多态性方法(PCR-RFLP)进行CYP7A1基因A-204C基因座Eco31I酶切多态性分析。结果可见CYP7A1基因A-204C多态基因座等位基因C、A频率在CHD组和正常对照组分别为0.840、0.160和0.822、0.178, 基因频率分布符合Hardy-Weiberg平衡定律。CYP7A1基因A-204C多态基因座基因型频率, 等位基因A、C频率在CHD患者组和正常对照组间比较差异无显著性(P>0.05), 但冠心病患者组中AA, AC, CC三种基因型之间总胆固醇(TC)的差异具有显著性(P<0.05), AA基因型患者的高密度脂蛋白胆固醇(HDL-C)和低密度脂蛋白胆固醇(LDL-C)水平较AC, CC基因型患者显著降低(P<0.05); 而对照组中CC, CA基因型个体间总胆固醇(TC)水平差异有显著性(P<0.05)。中国汉族与白种人CYP7A1基因A-204C多态基因座等位基因频率比较, 两者差异具有显著性(P<0.01)。提示CYP7A1基因A-204C多态基因座多态性与CHD无相关性, 但与总胆固醇存在较密切的关联(P<0.05), 在CHD患者组AA基因型中高密度脂蛋白胆固醇、低密度脂蛋白胆固醇水平显著下降(P<0.05)。Abstract: To study the distribution of Eco31I restriction polymorphism in nucleotide -204 of 7 α -hydroxylase gene(CYP7A1) in Sichuan Han population of China and association of the polymorphism with coronary heart disease(CHD), CYP7A1 genotyping was performed by using PCR-RFLP approach in 183 CHD patients and 101 control subjects. 7 α -hydroxylase gene allele frequencies of C, A were 0.840 and 0.160 in CHD group and 0.822 and 0.178 in control group, respectively. There was no significant difference in frequencies of allele and genotypes in A-204C polymorphism between CHD group and control group (P>0.05). However, in CHD patients there was significant difference in total cholesterol (TC) levels among CC, CA and AA genotypes (P<0.05), and the levels of high density lipoprotein cholesterol (HDL-C) and low density lipoprotein cholesterol (LDL-C) in CHD patients with AA genotype were lower than those in CHD patients with CC and CA genotypes (P<0.05). In control group there was significant difference in TC levels between CC and CA genotypes (P<0.05). The frequencies of C, A alleles at A-204C polymorphic site were significantly different from those reported in white people (P<0.05). The results indicating that no direct association was found between the A-204C polymorphism and CHD, but there was significant correlation between this polymorphism and the levels of TC, and there was significant correlation in CHD patient group between this polymorphism and levels of HDL-C and LDL-C.

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