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研究论文

温州地区2型糖尿病患者线粒体DNA 3243、3316位点的突变筛查

赵 晶[1], 季敬璋[1], 汪大望[2], 张 洁[1], 吴惠洁[3], 吕建新[1]

1. 温州医学院检验医学院, 浙江省医学遗传学重点实验室, 温州 325035; 2. 温州医学院附属第一医院内分泌科, 温州 325000; 3. 温州医学院附属第一医院八院区检验科, 温州 325000

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摘要 为了解浙江省温州地区2型糖尿病病人中线粒体DNA tRNALeu (UUR)基因A3243G及NADH 脱氢酶亚单位1 (ND1)基因G3316A位点突变的发生频率, 并探讨突变与2型糖尿病主要临床指标出现的相关性。对随机收集的无血缘关系的244例温州地区2型糖尿病患者进行研究, 同时选择156例无 DM 家族史的糖耐量正常者作为对照组, 用聚合酶链反应及限制性片段长度多态性分析技术进行点突变筛选, 筛选到的异质性突变样本经T-A克隆后再作测序和变性高效液相色谱(DHPLC)确证。结果在244例的2型糖尿病患者中检出A3243G突变1例(0.410%), 156例对照者中未检出该突变, 突变发生率在两组间差异无统计学意义(P>0.05); 2型糖尿病患者中检出G3316A突变4例(1.639%), 156例对照者中检出突变2例(1. 282%), 突变发生率在两组间差异无统计学意义(P>0.05)。结果表明线粒 体tRNALeu (UUR) 基因A3243G突变在浙江温州2型糖尿病人群中发生频率低, 不是温州人群中2型糖尿病的常见病因。线粒体ND1基因G3316A突变在糖尿病人群中的发生频率也较低, 且在正常人群中也有出现, 可能仅为人群中线粒体DNA的基因多态性。

关键词 2型糖尿病 线粒体DNA 点突变 多态性

分类号 R394

Detecting of mtDNA Mutations at Position A3243G and G3316A in Patients with type 2 Diabetes Mellitus in Wenzhou

ZHAO Jing[1], JI Jing-Zhang[1], WANG Da-Wang[2], ZHANG Jie[1], WU Hui-Jie[3], LU Jian-Xin[1]

1. School of Laboratory Medicine, Wenzhou Medical College, Zhejiang Provincial Key Laboratory of Medical Genetics, Wenzhou 325035, China; 2. Department of Endocrinology, the First Affiliated Hospital of Wenzhou Medical College, Wenzhou 325000, China; 3. The Eighth Department Clinical Laboratory,the First Affiliated Hospital of Wenzhou Medical College, Wenzhou 325000, China

Abstract

Abstract: To investigate the frequencies of mitochondria DNA (mtDNA) tRNALeu (UUR) point mutation A3243G and NADH dehydronase subunit 1(ND1) gene point mutation G3316A in Wenzhou area of Zhejiang Province, and to explore the correlation between these mutations and the clinical manifestations in patients with type 2 mellitus diabetes(T2DM). Two hundreds and forty-four unrelated patients with T2DM and 156 healthy subjects without family history of T2DM were enrolled in Wenzhou area in this study and screened for the point mutations mentioned above with polymerase chain reaction (PCR) and restricted fragment length polymorphism(RFLP) analysis. The heterogeneous mutations were confirmed with DNA sequencing and denaturing high performance liquid chromatography (DHPLC) following T-A cloning of PCR products. The percentage of A3243G mutation in group of patients with T2DM and control were 0.410% and 0.0% (1/244 vs 0/156), respectively; however, there's not any significant difference between these two groups in frequency of A3243G mutation (P>0.05). G3316A mutation was detected in 4 of 244 cases with T2DM (1.639%) and 2 of 156 healthy controls (1. 282%), showing that there's also no statistic difference between these two groups in frequency of G3316A mutation (P>0.05). It's shown that the frequency of mtDNA tRNALeu (UUR) A3243G mutation is fairly low in patients with T2DM in Wenzhou area. Thus it's reasonable to assume that this mutation may not be involved in the development and progression of T2DM. Furthermore, it's demonstrated that the rate of G3316A mutation of mtDNA ND1 gene is rare in patients with T2DM in Wenzhou area and this mutation also happened in healthy control. It's suggested that G3316A mutation is just a gene polymorphism of mtDNA and not related to the pathogenesis of T2DM.

Key words type 2 diabetes mellitus mitochondrial DNA point mutation polymorphism

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