遗传　1997 19 (2): 5-8　ISSN: 0379-4172　CN: 11-5450/R

线粒体基因突变与NIDDM发生的关系 Mitochondrial DNA Mutation Associated with NIDDM

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收稿日期　修回日期　网络版发布日期　接受日期

摘要　采用PCR-SSCP、PCR-RFLP及PCR产物直接测序等技术对90例NIDDM(即非胰岛素依赖型糖尿病)及80例正常对照个体的血细胞线粒体DNA进行了突变分析。结果在2例患者中发现线粒体DNA(mitochondrial DNA,mtDNA) ND1(NaDH Dehydrogenase subunit Ⅰ)基因上3316位点存在G→A的点突变,导致丙氨酸错义突变成苏氨酸,而在80例正常对照个体中均不存在此位点突变。国内外已证实的和1.5%NIDDM发生有关的mtDNA tRNA Leu^(UUR)基因上3243位点A→G的突变在本实验中并未发现。由此推断,3316位点G→A的突变可能与NIDDM的发生在关,3243位点A→G的突变率确实很低,可见糖尿病的发生在线粒体遗传上具有广泛的异质性。

Abstract:Using PCR-SSCP,PCR-RFLP and PCR product direct sequencing techniques,we analysed the mitochondrial DNAs(mtDNAs)of 90 patients with NIDDM (Non Insulin-Dependent Diabetes Mellitus)and those of 80 normal controls.The results showed that a G to A mutation which leads alanine's missence mutaton to threonine in the mitochondrial ND1(NaDH Dehydrogenase subunit I) gene at nucleotide pair 3316 occurred in the blood cells of 2 patients.We have not however,indentified with the A to G mutation at nucleotide pair 3243 of the mitochondrial tRNA Leu(UUR) gene,which has been reported to associate with NIDDM in about 1.5% of the diabetic population.We infer that the mutation at position 3316 is perhaps associated with the development of NIDDM,the occurance of the mutation at position 3243 is actually rare,and NIDDM has an intensive mitochondrial genetic heterogenous background.

关键词　非胰岛素依赖型糖尿病　线粒体DNA　错义突变　Key words　Non-Insulin dependent diabetes mellitus (NIDDM)　Mitochondrial DNA(mtDNA)　Missence mutation
分类号

Abstract

Key words

DOI:

通讯作者