

研究报告

一个2型糖尿病家系中新发现的线粒体DNA G7444A 突变分析

程祖建, 杨滨, 刘奇才, 江凌, 谢海花, 欧启水

福建医科大学附属第一医院检验科, 福建医科大学基因诊断研究室, 福建医科大学医学检验系, 福州 350005

收稿日期 2006-7-17 修回日期 2006-9-22 网络版发布日期 2007-3-12 接受日期

摘要

应用PCR-RFLP和测序对一个2型糖尿病家系的线粒体DNA G7444A的突变进行检测, 并分析其临床资料的特点。结果发现, 27例家系成员中, 11例母系亲属均存在线粒体DNA G7444A突变, 而配偶及父系亲属中未发现该突变。11例突变者中确诊为2型糖尿病患者5例, 糖耐量受损1例, 均表现为乳酸和血糖增高。因此, 线粒体DNA G7444A突变是该家系中糖尿病的遗传易感因素, 是导致2型糖尿病的一个新的突变位点。

关键词 [线粒体DNA](#) [2型糖尿病](#) [G7444A突变](#)

分类号

Study on a new point mutation of nt7444 G→A of mitochondrial DNA in a type 2 diabetes mellitus family

CHENG Zu-Jian, YANG Bin, LIU Qi-Cai, JIANG Ling, XIE Hai-Hua, OU Qi-Shui

Department of Laboratory Medicine, The First Affiliated Hospital, Fujian Medical University, Fuzhou 350005, China

Abstract

<P>Polymerase chain reaction restriction fragment length polymorphism (PCR-RFLP) and direct sequencing were applied to detect a new point mutation of nt7444G→A in the mitochondrial DNA in a type 2 diabetes mellitus family. The related clinical data were also collected and analyzed. mtDNA G7444A mutation in the cytochrome c oxidase I (COI) gene was found in 11 of 27 cases, all of whom were from the maternal side. Among them, 5 were confirmed to have type 2 diabetes mellitus, and one had impaired glucose tolerance. We conclude that the novel point mutation of mtDNA G7444A may be an independent factor associated with type 2 diabetes mellitus.</P>

Key words [mitochondrial DNA](#) [type 2 diabetes mellitus](#) [nt7444 G→A mutation](#)

DOI: 10.1360/yc-007-0433

通讯作者 欧启水 ouqishui@163.com

扩展功能	
本文信息	
▶ Supporting info	
▶ PDF(0KB)	
▶ [HTML全文](0KB)	
▶ 参考文献	
服务与反馈	
▶ 把本文推荐给朋友	
▶ 加入我的书架	
▶ 加入引用管理器	
▶ 复制索引	
▶ Email Alert	
▶ 文章反馈	
▶ 浏览反馈信息	
相关信息	
▶ 本刊中 包含“线粒体DNA”的相关文章	
▶ 本文作者相关文章	
· 程祖建	
· 杨滨	
· 刘奇才	
· 江凌	
· 谢海花	
· 欧启水	