遗传　2003 25 (3): 267-270　ISSN: 0379-4172　CN: 11-5450/R

Leber遗传性视神经病变家系的线粒体基因突变分析 Analysis of Mitochondrial Gene Mutations in Pedigrees of Leber's Hereditary Optic Neuropathy

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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　为探讨Leber遗传性视神经病变（Leber′s hereditary optic neuropathy，LHON）家系线粒体DNA（mtDNA）常见致病原发突变的频谱,用聚合酶链反应（polymerase chain reaction，PCR）和单链构象多态性（single-stranded conformational polymorphism，SSCP）以及DNA测序的方法，对13个家系22位临床诊断为LHON的患者及其母系亲属21人的线粒体DNA进行检测，同时检测71例正常人作为对照。临床拟诊为LHON的13个家系中，11个家系存在mtDNA位点11778 G→A突变，另2个家系存在14484位点T→C突变。说明中国LHON病人存在线粒体DNA 11778或14484位点突变，其中14484位点突变在国内尚未见报道。

Abstract:The purpose of the study is to investigate the frequency of common pathogenic primary mitochondrial DNA mutations in pedigrees of Leber′s hereditary optic neuropathy (LHON).Mutations were determined by polymerase chain reaction (PCR),single-stranded conformational polymorphism (SSCP) and DNA sequencing.Twenty-two patients with suspicion of LHON and twenty-one their maternal relatives underwent molecular genetic evaluation.Seventy-one normal individuals underwent molecular genetic evaluation as control at the same time.Members from 13 families with suspicion of LHON,11 families had nucleotide position nt11778 G→A mutations.Another 2 families had nt14484 T→C mutations.It is concluded that the point mutations at nucleotides 11778 and 14484 are primary LHON mutations,but the point mutation of nt14484 is rare in Chinese.

关键词　Leber遗传性视神经病　线粒体DNA　点突变　PCR-SSCP Key words　Leber′s hereditary optic neuropathy　mitochondrial DNA　point mutation　PCR-SSCP

分类号

Abstract

Key words

DOI:

通讯作者