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研究报告

近亲结婚所致一遗传性非综合征型耳聋家系的调查

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摘要 耳聋是一种最常见的人类感觉系统缺陷，在已发现的遗传性耳聋中，有70%的属于非综合征型听力缺损。据估计非综合征型遗传性耳聋基因总数在100个以上, 目前已经确定了近80个非综合征型遗传性耳聋的遗传位点, 其中23个基因已经被成功克隆。文章报道一遗传性非综合征型耳聋家系。该家系中存在2代近亲结婚，共2代13人出现聋哑症状。经遗传分析，该家系的遗传方式与常染色体显性或隐性遗传均不符合，提示此家系中的非综合征型遗传性耳聋可能为线粒体突变所致。

关键词 非综合征型耳聋；近亲婚配；线粒体突变

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A Family with Nonsyndromic Hearing Impairment Caused by Intermarry

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Abstract

Deafness is the most prevalent sensory system impairment in human, about 70 % of genetic deafness belongs to nonsyndromic hearing impairment. It was estimated that the total number of genes involved in nonsyndromic hereditary deafness was over 100. So far, approximate 80 loci have been mapped to human chromosome, and 23 genes have been identified. In this paper, a family with nonsyndromic hearing impairment caused by intermarry was reported. There were 13 sufferers in two generations. Deduced from genetic analysis, neither autosomal dominant nor autosomal recessive inheritance was identified in this family, which suggested that hearing impairment in the family was probably caused by mitochondrial mutations.

Key words Nonyndromic hearing impairment intermarry mitochondrial mutations

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