

25个携带线粒体12S rRNA A1555G突变的中国汉族非综合征型耳聋家系

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摘要 线粒体12S rRNA A1555G突变是引起氨基糖甙类药物诱导的非综合征型耳聋的重要原因之一。文章对收集的25个携带A1555G突变的中国汉族非综合征型耳聋家系进行了临床和分子遗传学评估。结果表明, 这25个家系的母系成员在耳聋外显率、听力损失严重程度和发病年龄上存在较大差异。当包括和不包括氨基糖甙类药物使用史时, 耳聋的平均外显率分别为28.1%和21.5%, 排除氨基糖甙类药物时, 耳聋的平均发病年龄从1~15岁不等。线粒体全序列分析发现了16个新变异, 不同的线粒体DNA多态性位点显示这25个家系分别属于东亚人群A、B、D、F、G、M、N和R单倍型, 其中线粒体单倍型B的家系耳聋外显率和表现度较其他单倍型高。此外, 7个继发突变位点和21个高保守性位点突变可能增加了这些家系的耳聋外显率。*GJB2*基因上未检测到与耳聋相关的突变, 表明在本研究的耳聋家系中, *GJB2*基因可能没有参与A1555G突变的表型表达。以上各方面提示, 线粒体单倍型和其他因素可能参与了这25个家系耳聋患者的表型修饰。

关键词: 线粒体12S rRNA A1555G突变 非综合征型耳聋 氨基糖甙类抗生素 单倍型

Abstract: Mitochondrial 12S rRNA A1555AG mutation is one of the important causes of aminoglycoside-induced and nonsyndromic deafness. We report here the clinical, genetic and molecular characterization of 25 Chinese families carrying the A1555G mutation. Clinical and genetic characterizations of these Chinese families exhibited a wide range of penetrance, severity and age-at-onset of hearing impairment. The average penetrances of deafness were 28.1% and 21.5%, respectively, when aminoglycoside-induced hearing loss was included or excluded. Furthermore, the average age-of-onset for deafness without aminoglycoside exposure ranged from 1 and 15 years old. Their mitochondrial genomes exhibited distinct sets of polymorphisms including 16 novel variants, belonging to ten Eastern Asian haplogroups A, B, D, F, G, M, N and R, respectively. Strikingly, these Chinese families carrying mitochondrial haplogroup B exhibited higher penetrance and expressivity of hearing loss. In addition, 7 known secondary mutations and 21 variants resided at the highly conservative residues may enhance the penetrance of hearing loss in these Chinese families. Moreover, the absence of mutation in *GJB2* gene suggested that *GJB2* may not be a modifier for the phenotypic expression of the A1555G mutation in these Chinese families. These observations suggested that mitochondrial haplotypes and other modifiers may modulate the variable penetrance and expressivity of deafness among these Chinese families.

Keywords: mitochondrial 12S rRNA, A1555G mutation, nonsyndromic hearing loss, aminoglycoside antibiotics, haplogroup

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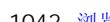
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