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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

彭光华[1,2], 郑斌娇[3], 方芳[3], 伍越[3], 梁玲芝[3], 郑静[3,5], 南奔宇[1], 余啸[1], 唐霄雯[3], 朱翌[4], 吕建新[3], 陈波蓓[1], 管敏鑫[3,5]

1. 温州医学院附属第二医院 附属育英儿童医院耳鼻喉科, 温州 325027; 2. 余姚市人民医院耳鼻咽喉科, 宁波 315400; 3. 温州医学院Attardi线粒体生物医学研究院和浙江省医学遗传学重点实验室, 温州 325035; 4. 温州医学院附属第一医院耳鼻喉科, 温州 325027; 5. 浙江大学生命科学学院, 杭州 310058

PENG Guang-Hua[1,2], ZHENG Bin-Jiao[3], FANG Fang[3], WU Yue[3], LIANG Ling-Zhi[3], ZHENG Jing[3,5], NAN Ben-Yu[1], YU Xiao[1],TANG Xiao-Wen[3], ZHU Yi[4], LU Jian-Xin[3], CHEN Bo-Bei[1], GUAN Min-Xin[3,5]

1. Department of Otolaryngology, the Second Affiliated Hospital & Yuying Children ' s Hospital of Wenzhou Medical College, Wenzhou 325027, China; 2. Department of Otolaryngology, Yuyao People ' s Hospital, Ningbo 315400, China; 3. Attardi Institute of Mitochondrial Biomedicine and Zhejiang Provincial Key Laboratory of Medical Genetics, School of Life Sciences, Wenzhou Medical College, Wenzhou 325035, China; 4. Department of Otolaryngology, the First Affiliated Hospital of Wenzhou Medical College, Wenzhou 325027, China; 5. College of Life Sciences, Zhejiang University, Hangzhou 310058, China

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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 cilinical, 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, respec-tively. 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 penetrace 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 pene-trance and expressivity of deafness among these Chinese families.

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

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通讯作者 管敏鑫 Email: gminxin88@gmail.com

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