

## 胞质异质性——人类肿瘤组织线粒体基因突变的普遍现象

# Heteroplasmy: A Common Phenomenon of Mitochondrial Genome Mutations in Human Tumor Tissues

谭端军<sup>1</sup>, 刘玲玲<sup>1</sup>, 文毅<sup>1</sup>, 刘鹏<sup>1</sup>, Julia Chang<sup>2</sup>, Kun-Tu Yeh<sup>2</sup>, LeeJun C Wong<sup>3</sup> TAN Duan-Jun<sup>1</sup>, LIU Ling-Ling<sup>1</sup>, WEN Yi<sup>1</sup>, LIU Peng<sup>1</sup>, Julia Chang<sup>2</sup>, Kun-Tu Yeh<sup>2</sup>, LeeJun C Wong<sup>3</sup>

1.解放军总医院老年心血管病研究所, 北京 100853; 2.彰化基督教医院病理科, 彰化, 中国台湾 500; 3.乔治城大学医学中心分子与人类遗传研究所, 华盛顿,美国 20007 1. Institute of Geriatric Cardiology of Chinese PLA General Hospital, Beijing 100853,China; 2. Department of Pathology, Changhua Christian Hospital, Changhua, China Taiwan 500; 3.Institute for Molecular and Human Genetics, Georgetown University Medical Center, Washington DC 20007, USA

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### 摘要

为了探讨不同肿瘤组织中线粒体基因体细胞性突变的胞质异质性和同质性状态, 利用32对重叠引物对149例肿瘤组织和匹配的正常组织的全线粒体基因进行PCR扩增, 并同时进行时相温度梯度凝胶电泳扫描突变筛选, 基因测序确定突变类型与异质状况。结果表明, 不同肿瘤组织中线粒体基因体细胞性突变的异质率不同, 口腔癌(65%)和食道癌(64%)具有较高的异质率, 其次为乳腺癌(45.9%)。4种转换形式的发生频率Hm→Hm > Hm→Ht > Ht→Hm > Ht→Ht。碱基转换的主要转换形式为Hm→Hm, 碱基颠换则以Hm→Ht。认为胞质异质性是人类肿瘤组织线粒体基因突变的普遍现象。Abstract: To explore the status of heteroplasmy and homoplasmy of Mitochondrial DNA somatic mutations in different tumors. DNA from 149 tumors and corresponding normal tissues were extracted and entire mitochondrial genome was amplified using 32 pairs of overlapping primers. The somatic mutations were screened by temporal temperature gradient gel electrophoresis and their heteroplasmic statute were identified by sequencing. The results showed that the incidence rate of heteroplasmy of mitochondrial DNA somatic mutations varies in different tumors. There is a high rate of heteroplasmic mutation in oral cancer (65%) and esophageal cancer (64%), followed by breast cancer (45%). The frequency of four transfer types is Hm (homoplasmy)→Hm (heteroplasmy) > Hm→Ht > Ht→Hm > Ht→Ht. The main transfer forms of transition and transversion mutations are Hm→Hm and Hm→Ht respectively. Heteroplasmy is a common phenomenon in mitochondrial DNA somatic mutations of human tumors.

关键词 [线粒体基因](#) [突变](#) [异质性](#) [同质性](#) [肿瘤](#) Key words [Mitochondrial genome](#) [mutation](#) [heteroplasmy](#) [homoplasmy](#) [tumor](#)

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### Abstract

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