

论著

# 潮汕地区食管鳞状细胞癌患者与正常人群O6-甲基鸟嘌呤-DNA甲基转移酶基因多态性的初步分析

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**摘要** 背景与目的: 初步研究中国食管癌高发区之一潮汕地区食管癌患者和正常人群O6-甲基鸟嘌呤-DNA甲基转移酶(MGMT)基因编码84、143及160位密码子的单核苷酸多态性(Single nucleotide polymorphism, SNP)。材料与方法: 应用双色荧光杂交微阵列技术分别检测潮汕地区100例食管鳞状细胞癌患者及65例健康对照外周血MGMT基因84位密码子(C2740214T)单核苷酸多态性; 用同样的方法检测76例食管鳞状细胞癌患者和50例健康对照MGMT基因143位密码子(A2798995G)、160位密码子(G2799046A)单核苷酸多态性。结果: 基因型分布符合Hardy - Weinberg 定律。统计分析显示食管鳞状细胞癌组与对照组84位密码子2740214C和2740214T等位基因频率差异无统计学意义(P=0.402), 2740214CT基因型在病例组和对照组中所占的比例分别为16%和12.3%, 2740214TT基因型在病例组中所占的比例是2%。食管鳞状细胞癌组变异基因型拥有者(CT+TT)高于对照组, 但差异无统计学意义(P=0.327), 2740214TT基因型仅见于个别肿瘤患者, 而在正常对照中未发现。143位密码子A2798995G位点仅在对照组中发现一例杂合子, 其余均为野生型纯合子, 病例组与对照组基因型和等位基因频率差异均无统计学意义。160位密码子G2799046A位点检测在所有的病人和对照中均为野生型纯合子。结论: 潮汕地区食管鳞状细胞癌患者和正常人群中存在MGMT基因多态性, 84位密码子C2740214T位点的突变型纯合子仅见于肿瘤患者, 可能与食管鳞状细胞癌有关。

**关键词** [O6-甲基鸟嘌呤-DNA甲基转移酶](#); [单核苷酸多态性](#); [杂交](#); [食管癌](#)

## Polymorphisms of O6-Methylguan-ine-DNA Methyl-transferase Gene in Chinese Chaoshan Esophageal Cancer Patients

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**Abstract** **BACKGROUND & AIM:** To study the polymorphisms of the O6-Methylguanine-DNA methyltransferase (MGMT) gene codon 84(C2740214T), codon 143(A2798995G) and codon 160 (G2799046A) for esophageal cancer and to assess the prevalence of these polymorphisms in Chinese Chaoshan population. **MATERIAL AND METHODS:** A microarray-based method for genotype analysis of esophageal cancer patients and healthy controls were performed. Amplified PCR products from genomic DNA specimens were spotted and immobilized onto glass slide to fabricate a microarray, then detected by hybridization with dual-color probes. 100 patients and 65 healthy controls were tested for codon 84. 76 patients and 50 healthy controls were tested for codons 143 and 160. **RESULTS:** The prevalence of the 2740214 CT in patients was 0.16 and 0.123 in controls. The prevalence of the 2740214 TT was 0.02 in patients but not detected in controls. Heterozygote at codon 143 was found in one healthy individual, but didn't exist in the cancer patients. In all the samples, no mutation was detected at codon 160. **CONCLUSION:** These results suggested that the polymorphisms of MGMT exist in Chinese Chaoshan population. Homozygous mutant at codon 84 might be correlated with carcinogenesis.

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