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简报

多重连接依赖的探针扩增技术检测中国人遗传性非息肉病性结直肠癌错配修复基因大片段缺失

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摘要 摘要：目的 了解中国人遗传性非息肉病性结直肠癌（HNPCC）家系hMSH2和hMLH1基因大片段缺失特点。方法 采用多重连接依赖的探针扩增（MLPA）技术和GeneMapper分析技术检测17个HNPCC家系先证者hMSH2和hMLH1基因种系大片段缺失。结果 在3个家系中分别发现hMSH2基因第8外显子、1～6外显子和1～7外显子3种大片段缺失类型, 未发现hMLH1基因大片段缺失。大片段缺失占hMSH2和hMLH1基因总种系病理性突变的19%。结论 中国人HNPCC错配修复（MMR）基因大片段缺失发生率较高, hMSH2基因缺失可能更为常见。在分子遗传学检测中有必要开展MMR基因大片段缺失的检测。

关键词 遗传性非息肉病性结直肠癌 大片段缺失 多重连接依赖的探针扩增

分类号

Detection of Large Intragenic Mismatch Repair Genes Deletions in Chinese ereditary Nonpolyposis Colorectal Cancer Families with Multiplex Ligation-dependent Probe Amplification Technique

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Abstract ABSTRACT:Objective To gain an insight into the large intragenic hMSH2 and hMLH1 deletions in Chinese hereditary nonpolyposis colorectal cancer（HNPCC）families. Method The large intragenic hMSH2 and hMLH1 deletions in 17 probands of HNPCC families were detected with multiplex ligation-dependent probe amplification（MLPA）and GeneMapper techniques. Results Three large intragenic hMSH2 deletions of exon 8, exon 1-6, and exon 1-7 were found in three families respectively, and no hMLH1 deletion was found. The deletions accounted for 19% of the total hMSH2 and hMLH1 germline pathogenic mutations. Conclusions The incidence of large intragenic mismatch repair（MMR） genes deletions is relatively higher in Chinese families, and hMSH2 deletions may be more common. It is necessary to detect the large intragenic MMR genes deletions in the molecular detection of HNPCC.

Key words hereditary nonpolyposis colorectal cancer large intragenic deletion multiplex ligation-dependent probe amplification

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