

论著

骨性关节炎患者关节软骨hMLH1启动子区甲基化水平

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摘要: 目的: 通过对骨性关节炎(osteoarthritis, OA)患者关节软骨人类错配修复基因1(human MutL homologue1, hMLH1)启动子区甲基化状态及hMLH1蛋白表达的检测, 探讨hMLH1启动子区甲基化在OA发生发展中的作用。方法: 采用亚硫酸氢钠法处理基因组DNA, 甲基化特异性PCR检测hMLH1基因启动子甲基化情况; 免疫组织化学方法检测hMLH1蛋白的表达。结果: OA患者组关节软骨hMLH1启动子区甲基化阳性率明显高于健康人对照组($\chi^2=30.634, P<0.001$); OA患者组关节软骨hMLH1蛋白表达率明显低于健康人对照组($\chi^2=37.724, P<0.001$); OA患者组关节软骨hMLH1启动子区启动子甲基化与蛋白表达呈显著负相关($r_s=-0.554, P<0.001$)。结论: OA患者组关节软骨hMLH1启动子区发生了甲基化。其高甲基化状态影响了hMLH1蛋白的表达, 可能参与OA疾病的发生发展。

关键词: 骨性关节炎 DNA 甲基化 hMLH1 启动子 甲基化特异性PCR

hMLH1 gene promoter methylation in joint cartilage in patients with osteoarthritis

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Abstract: Objective: To investigate the role of Human MutL homologue 1 (hMLH1) gene promoter methylation in the occurrence and development of osteoarthritis (OA).
Methods: General DNA was dealt with sodium bisulfite. The methylation of hMLH1 promoter was detected by methylation-specific PCR (MSP). hMLH1 protein expression in joint cartilage was detected by immunohistochemical method.
Results: The positive percent of hMLH1 promoter methylation in OA patients was higher than that in healthy persons ($\chi^2=30.634, P<0.001$); the positive percent of hMLH1 protein in OA patients was significantly lower than that in healthy persons ($\chi^2=37.724, P<0.001$); promoter methylation and protein expression level of hMLH1 gene showed negative correlation ($r_s=-0.554, P<0.001$).
Conclusion: hMLH1 promoter is hypermethylated in joint cartilage cells of OA patients. Hypermethylation may affect the protein expression of hMLH1, which might play a role in the occurrence and development of OA.

Keywords: osteoarthritis DNA methylation hMLH1 promoter MSP

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