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摘要：

目的：探讨散发性结直肠癌微卫星不稳定(microsatellite instability，MSI)情况及其与错配修复 (mismatch repair，MMR) 蛋白MLH1、MSH2、MSH6、PMS2表达缺失的相关性，并总结MSI散发性结直肠癌的临床病理学特征。方法：多重荧光PCR法检测散发性结直肠癌肿瘤组织DNA的微卫星不稳定性，免疫组化(Immunohistochemistry，IHC)S-P法检测散发性结直肠癌肿瘤组织MLH1、MSH2、MSH6、PMS2蛋白的表达缺失，分析MSI发生与MMR蛋白表达缺失及临床病理特征的相关性。结果：75 例散发性结直肠癌检出MSI 21例(28%)，包括 MSI-H 19例、MSI-L 2例，其他54例(72%)为MSS。检出MMR蛋白表达缺失16例(21.33%)，其中15例(93.75%)为MSI-H、1例(6.25%)为MSS；MMR蛋白表达59例(78.67%)，其中4例(6.78%)为MSI-H、2例(3.39%)MSI-L，其他53例为MSS。MSI组MMR蛋白缺失率(15/21，71.43%)显著高于MSS组(1/54 ，1.9%) (P<0.01)。MSI与患者年龄，是否黏液腺癌，肿瘤有远处转移有关 (P<0.01)，其中MSI-H好发于年龄>50岁、肿瘤远处转移、MMR蛋白缺失人群，且类型以黏液腺癌为主。结论：散发性结直肠癌肿瘤组织中MSI发生率高于MMR蛋白缺失率，并且MSI-H的散发性结直肠癌转移风险较低、预后较好。检测MSI 状态对提高结直肠癌的预防、诊断和治疗水平，降低结直肠癌的发病率和病死率有着重要意义。

关键词：散发性结直肠癌 微卫星不稳定 错配修复蛋白 免疫组织化学 临床病理学特征

The relationship of microsatellite instability state with loss of mismatch repair proteins and clinical pathological characteristics in sporadic colorectal cancers Download Fulltext

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Abstract:

Objective : To investigate the relationship of microsatellite instability (MSI) condition with the deletion of mismatch repair (MMR) proteins (MLH1, MSH2, MSH6, PMS2) and the clinical pathological characteristics in sporadic colorectal cancers. Methods: MSI of DAN samples from sporadic colorectal cancers was examined with multiplex fluorescent PCR. MMR proteins in sporadic colorectal cancer tissues were detected with SP immunohistochemistry (IHC) method. Results: In 75 cases of sporadic colorectal cancers, 21 cases (28%) had MSI, including MSI-H (19 cases) and MSI-L (2). MMR protein deletion was found in 16 cases (21.3%), of which 15 cases (93.75%) were MSI-H, and the other case (6.2%) was MSS. The rate of MMR protein deletion in MSI group (15/21 , 71.43%) was significantly higher than that of MSS group (1/54 , 1.9%, P<0.01). Notably, 4 cases with MSI-H (6.78%) and 2 cases (3 39%) with MSI-L did not have any NMR protein deletion. MSI in these patients was associated with age, mucus content of the adenocarcinoma, and tumor metastasis (P<0.01). MSI-H tends to occur in >50 years old individuals who have mucinous adenocarcinoma harboring MMR protein deletion and often have no distant metastasis of the cancer. Conclusion: The incidence of MSI is higher than that of MMR protein deletion in sporadic colorectal cancer. These with MSI-H have lower risk of metastasis and better prognosis. Therefore, detecting MSI is valuable for improving the prevention, diagnosis, and treatment of sporadic colorectal cancer, and for reducing its incidence and mortality.

Keywords:sporadic colorectal cancer microsatellite instability mismatch repair protein immunohistochemistry characteristics of clinical pathology

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