

Immunohistochemical screening of hMLH1 and hMSH2 gene mutations in patients diagnosed with colorectal cancer and microsatellite instability suspicion

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Study aim: immunohistochemical screening of hMLH1 and hMSH2 gene mutations in patients diagnosed with colorectal cancers, suspected of having microsatellite instability, as diagnosed between January 2002 and December 2009 in the Surgery Department of the CF Clinical Hospital Cluj-Napoca (prospective non-randomised study).

Methods: inclusion criteria were adenocarcinoma pathology finding and also minimum one of the revised Bethesda criteria for genetic testing of microsatellite instability in colorectal cancers. 110 eligible patients were divided in 2 study groups according to the number of Bethesda criteria met (group A – 1 criteria; group B – 2 or more criteria). Both groups were statistically compared considering the clinical and pathological parameters specific to the Lynch syndrome. We performed immunohistochemical staining to determine the expression of hMLH1 and hMSH2 genes in the tumors of all the patients.

Results: we found the differences in age, colorectal family history and right colon tumor site between the two groups to be statistically significant. Immunohistochemical stainings showed lack of hMLH1 gene expression in 9 patients and of hMSH2 gene in 4 patients respectively.

Conclusions: Immunohistochemical staining can identify patients who need to be genetically tested for mutations of the DNA mismatch repair genes, in order to establish the correct diagnostic of Lynch syndrome.

Key words: immunohistochemistry, colorectal cancer, micro-satellite instability, Lynch syndrome

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