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Analysis of the association of *APC*, *MLH1* and *RASSF1A* genes methylation with the risk of colorectal cancer

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Colorectal cancer (CRC) is the fourth most common cause of cancer morbidity and the 3rd most common cause of cancer mortality in Kazakhstan. The most important condition for successful treatment of CRC is the detection of tumor in the early stages. Diagnostic and prognostic value in solving this problem can have epigenetic methylation of key genes of colorectal carcinogenesis. The aim of this study was to investigate the association of methylation status of *APC*, *MLH1* and *RASSF1A* genes, involved in the regulation of cell cycle and apoptosis, with the risk of developing CRC. For study, a total 50 samples of tumor tissue from patients with sporadic CRC were collected, of which 16 patients simultaneously with biopsy of tumor tissue, samples of peripheral unaffected intestinal tissue and in 26-peripheral blood were taken. For the control group, peripheral blood samples of 30 conditionally healthy individuals were selected. DNA was isolated by phenol-chloroform extraction. To determine methylation status of the genes' promoter region, methyl-sensitive and methyl-specific PCR methods were used. Diagnostic characteristics of the test for methylation promoter sequence of *APC* and *RASSF1A* genes in the intestinal tissue to detect CRC are following: Sensitivity- 4.00% and 10.00%, respectively, and specificity- 100.00% for both genes. For CRC diagnostics sensitivity of the test used for *MLH1* gene promoter sequence methylation in the intestinal tissue and peripheral blood was 30.00% and 3.85%; specificity- 87.50% and 100.00%, respectively. Preliminary results show the high specificity of the tests for promoter methylation of *APC* and *RASSF1A* genes in the intestinal tissue, the *MLH1* gene- in the intestinal tissue and peripheral blood in CRC diagnostics. It is assumed further collection of clinical material and sampling extension for further study of the role of epigenetic changes of these genes in the development of CRC.

Biography

Zhunussova G has completed her PhD from Al-Farabi Kazakh National University. Title of her PhD thesis was: "Development of Genetic Markers Panel for the Screening of Familial and Sporadic Colorectal Cancer Cases in Populations from Kazakhstan". Now she is Head of Laboratory of Molecular Genetics, Institute of General Genetics and Cytology, Almaty, Kazakhstan. She has published more than 10 papers in reputed journals (*Tumor Biology* (IF 2.840), *Journal of Carcinogenesis and Mutagenesis* (IF 3.44), *South African Medical Journal* (IF - 1.712), and *Clinical Laboratory*, etc.). She is a Winner of the State Scientific Scholarship for Talented Young Scientists from MES of RK. Her research interests are human genetics, genetic aspects of carcinogenesis, genetic basis of multifactorial diseases, genetics of aging, molecular genetics of sporadic and hereditary (familial) colorectal cancer, and sports genetics.

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