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MINISATELITE INSTABILITY OF hMLH1, hMSH2 GENES IN HEREDITARY COLORECTAL ADENOMAS (LYNCH SINDROME)

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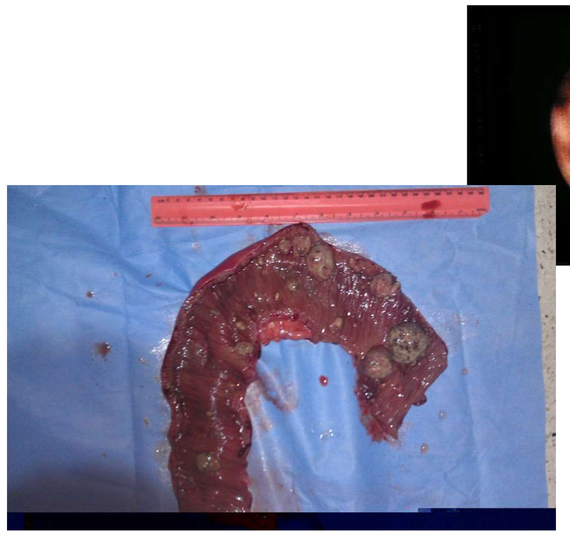
Introduction. In the light of the progress achieved in the area of improving the methods of recombined DNA technology, today it has become possible to diagnose a genetic disease, including the Lynch Syndrome as a form of epithelial colorectal neoplasia (ECRN) at the level of genes (hMLH1, hMSH2).

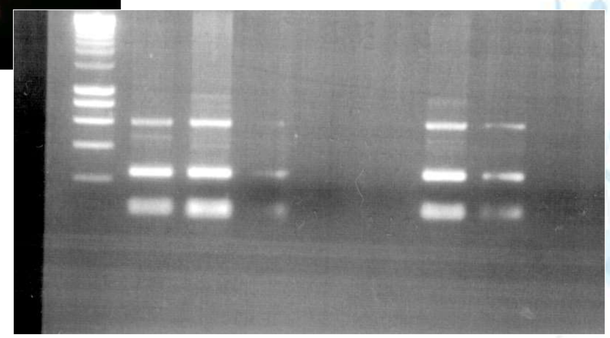
Keywords. Hereditary colorectal adenomas, genes (hMLH1, hMSH2).

Purpose. Analysis of the results of genetic research, at the molecular level of the hMLH1, hMSH2 genes involved in triggering the mechanism of tumorogenesis.

Material and methods. During the years 2012-2019, a group of 55 patients were observed and treated, of which 19 patients were diagnosed with IMS. In the identification of the genetic associations between the polymorphic DNA spectra and the clinical manifestations of the studied precancerous (tumorogenic) diseases, the PCR technique of the isolated DNA samples from the investigated patients was used.

Results. Following the RT-PCR reaction based on RNA isolated from the biological material, 30 cases (54.5%) with negative expression were found, a positive result of grade I (+) was determined in 14 cases (26, 2%) and grade II (++) in 10 cases (19%). The molecular-genetic origin of NECR was confirmed and the value of the polymorphic appearance of the hMLH1, hMSH2 genes, their level and frequency of expression in tumor tissues was determined.





Conclusions. The RT-PCR technique used in the experimental study contributes to increasing the efficiency of the medical-genetic consultation and to the eradication of colorectal cancer already at the early stages of development.