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TP53 and PIC3CA gene mutations as targets for circulating tumour DNA detection in colorectal and breast cancer patients

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Background. At the moment, liquid biopsy is at the peak of attention, challenging scientists with many questions. One of them is the determination of appropriate targets for ctDNA detection. A lot of variants have already been suggested for different localizations, but we decided to propose at once two equivalent targets for two different localizations in this study. We suppose that such an approach in future research can help to create a universal panel for detection and monitoring of different cancer types and, as a result, liquid biopsy could be made more available and convenient to general use in clinical practice.

Aim. Our study aimed to compare frequency of TP53 and PIC3CA mutations in patients with colorectal cancer (CRC) and breast cancer (BC) and determine their role in treatment monitoring of these cancers.

Methods and materials. 29 patients with histologically verified BC and 19 patients with CRC were screened for study participation. All of 48 patients underwent radical surgery for the underlying disease. Quantification of TP53 and PIC3CA gene mutations was performed using the QuantStudio 3D Digital PCR System (Applied Biosystems, USA). Both FFPE tumour tissue samples and patients' blood plasma samples taken before and after surgery were tested for the above mutations.

Results. Target mutations were detected in 78.9% of patients with CRC. 52.6% of CRC patients had mutation of TP53 gene, 10.5% – of PIK3CA, and 15.8% had both mutations simultaneously. In patients with BC, mutation of TP53 gene was detected in 20.7%, PIK3CA gene mutation – in 17.2%. The results of detecting mutations in FFPE samples were comparable with the results of plasma samples analysis. There was a significant decrease in ctDNA in patients' blood in the postoperative period, compared with the samples taken before surgery.

Conclusion. The obtained results show the significant importance of TP53 and PIK3CA gene mutations for CRC patients. In BC patients, these mutations appear less but also have a clinical significance. We suppose that mutations in the TP53 and PIK3CA genes, determined at different stages of treatment of patients with BC and CRC, can help to monitor the effectiveness of their treatment. Further research is needed for in-depth analysis.

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