

Next-Generation Sequencing and Molecular Diagnostic In Cancer †

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Abstract: The use of Next-Generation Sequencing in routine clinical molecular biology characterization of cancer is still insufficiently exploited in Romania. Moreover, NGS analysis of tumor specificity using the liquid biopsy as a starting point is a new and not fully characterized tool that cancer patients could benefit from worldwide. This data is part of a research project focused on the need to change the clinical practice in Romania by introducing liquid biopsy as a novel prognostic tool for personalized therapy in patients with advanced stages of cancer. DNA and RNA were isolated from plasma and tumor tissue samples of 17 patients diagnosed with advanced colon cancer. Until now, nucleic acid samples isolated from plasma belonging to 10 patients were sequenced using an NGS panel comprising 52 genes found to be frequently mutated in different cancer types. For 6 of these patients, NGS analysis of DNA and RNA isolated from tumor tissue was made. Results: The most frequent pathogenic variants found target hotspot regions in genes involved in the development and progression of colon cancer: TP53, KRAS, PIK3CA, APC. We also found some rare variants for this pathology: MAP2K1 mutations or EGFR CNVs. In conclusion, most of the variants detected in the tumor tissue were found in the corresponding plasma sample. Only the variants with low allelic frequency in the tumor were not detected in plasma. Some variants detected in plasma were not found in the tumor tissue, demonstrating tumor clonality.

Keywords: next-generation sequencing; liquid biopsy; colon cancer.

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Conflicts of Interest

The authors declare no conflict of interest.