



Fund “Nauka” Project № 25021 Resume – Competition-based Session 2025:

“Identification of genetic variants in miRNA genes with biomarker value in colorectal cancer through next-generation sequencing in a Bulgarian cohort”

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Colorectal carcinoma is the second most common malignant disease in women and the third most common in men. Identifying new biomarkers for early diagnosis, prognosis, and personalized treatment is a priority task in modern oncology.

The project aims to identify genetic variants in miRNA genes with biomarker value in Bulgarian patients with colorectal cancer through next-generation Nanopore sequencing (MinION Mk1D platform). Fifty samples (25 patients and 25 healthy controls) from previously studied cohorts will be analyzed.

The study includes isolation of genomic DNA, selection of target miRNA genes, preparation of sequencing libraries, and NGS sequencing. Bioinformatic processing will include variant identification, comparison with databases, and theoretical functional prediction. Statistical analysis will assess the association with disease susceptibility, correlation with clinicopathological parameters, survival, and biomarker potential. An algorithm for clinical prioritization of the identified variants will be developed.

Expected results include: genetic characterization of miRNA polymorphisms in a Bulgarian cohort, identification of new and population-specific variants, establishment of potential diagnostic and prognostic biomarkers, validated algorithm for prioritization of the identified polymorphisms in miRNA genes. The project will provide Nanopore sequencing technology and a trained research team.

The expected results from the implementation of this project proposal are divided into two categories:

1. Scientific research results

- Genetic characterization of miRNA genetic variants in CRC in a Bulgarian cohort;
- Identification of new/ unreported and population-specific polymorphisms;
- Identification of genetic variants with potential as diagnostic and prognostic biomarkers;
- Developing of algorithm for clinical prioritization of miRNA variants;

- Upon identification of innovative elements – patent application.

2. *Infrastructure results*

- Implementation of MinION Mk1D Nanopore sequencing technology in the Department of Biochemistry, Molecular Medicine, and Nutrigenomics, Medical University of Varna;
- Formation of a trained team with competencies in next-generation targeted sequencing technologies and bioinformatic data analysis.