TITLE: CLINICAL CHARACTERISTICS OF COLORECTAL CANCER IN PATIENTS DIAGNOSED WITH CHEK2 AND NOD2 GENE MUTATIONS.

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Introduction: Malignant neoplasms of the colon and rectum are among the most common cancers in humans. They are largely related to genetic predisposition. Some of the best-described genes for multi-organ predisposition to cancer are the CHEK2 and NOD2 genes. The purpose of this study was to validate existing clinical characteristics of colorectal carcinoma in CHEK2 and / or NOD2 mutation carriers, and especially 5-year survival rate.

Material and methods: The methodology of work was based on molecular and clinical analysis of 852 patients sequentially diagnosed for colorectal and rectum cancer treated in the Beskid Oncology Center in Bielsko Biała and Independent Public Clinical Hospital No. 2 PUM in Szczecin. The study group was analyzed for their clinical stage, the age when the disease started, 5-year survival rate and the presence of the CHEK2 mutation.

Results: Complete data was obtained from 553 people. 34 mutations in the CHEK2 gene and 57 mutations in the NOD2 gene were detected. There was no statistically significant trend for younger age at which the disease started, or a higher degree of clinical progression in patients with colorectal cancer and CHEK2 or NOD2 gene mutation. The largest difference in the risk of death between the patients at I and II stage, and the patients at III and IV stage of clinical progression was observed in CHEK 2 (OR-1.8, p-0.39) mutation carriers.

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