P074 Analysis of the frequency of occurrence of mutations in repair genes in patients with prostate cancer in the Republic of Bashkortostan

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Introduction & Objectives: Mutations in the HRR genes lead to disturbance of DNA repair, accumulation of errors, and increase genomic instability. The causes of deficiency can be both genetic (germinal and somatic mutations in the genes of the proteins involved in the process) and various epigenetic factors. The aim of the work: To determine the frequency of mutations in patients with metastatic and locally advanced prostate cancer in the Republic of Bashkortostan.

Materials & Methods: The analysis included 121 patients with prostate cancer living in the Republic of Bashkortostan from 2020 to 2022. Patients were surveyed in the developed bot-questionnaire for the Telegram messenger. The patient's blood and histological material were sent for HRR testing at the same time. Histological materials obtained less than 5 years ago were sent for the research.

Results: Molecular genetic analysis of venous blood and tumor tissue of oncological patients was carried out using NGS (next generation sequencing) research. The research group included germline and somatic mutations in repair genes. The results of the research in the region under consideration showed the territorial and national features of the presence of germline mutations in patients with prostate cancer. Based on the data obtained, the spectrum and the frequency of gene mutations, which are typical for the considered region, have been determined. As a result, the following mutations were identified with a characteristic frequency of occurrence: BRCA 1 - c.5266dupC, c.5382insC; BRCA 2 - c.1649_1650insTGTTCCTTACTATCCTTAGCCCTCTTT, c.3749dupA, c.1364C>A; RAD54L- c.2081_2082del; RAD51B - c.139C>T; ATM - c.3146T>A, c.9127delA, c.7792C>T; CHEK1: c.236G>A. According to the results of a patient survey, the presence of germline mutations in patients with burdened familial history was noted. In patients without a burdened familial history, when a mutation in the tumor tissue was detected, the germline mutation in the blood was not confirmed.

Conclusions: Using a questionnaire in the developed bot-questionnaire for the Telegram messenger allows to determine the risk group with germline mutations. The results of the research showed the territorial features of the frequency of occurrence of germline and somatic mutations in the Republic of Bashkortostan. This research allowed to determine the tactics of treatment among patients with prostate cancer.