Streszczenie w języku angielskim

Malignant epithelial tumors represent a significant health problem, requiring a multidisciplinary approach both in diagnostics and treatment. With the advancement of knowledge in the field of cancer genetics, the importance of molecular diagnostics in the detection, diagnosis, monitoring, and treatment of this group of diseases has significantly increased. This doctoral dissertation presents a series of publications covering the topic of the application of selected genetic diagnostic methods in the optimization of oncological management, with particular emphasis on bladder cancer diagnostics and the assessment of hereditary cancer predisposition syndromes. The first study evaluated the usefulness of whole-genome sequencing in relation to routinely used diagnostic methods for bladder cancer. The bioinformatic analysis of data obtained from sequencing DNA samples isolated from urine and peripheral blood allowed, among other things, the identification of characteristic genetic alterations specific to bladder cancer and potential markers for targeted therapies. The second study focused on the current state of knowledge on the significance of variants within the *TERT* gene promoter in detecting bladder cancer from urine samples. A retrospective analysis of data from the first publication of the series was also conducted, focusing on the presence of these variants and their potential utility in the early diagnosis of bladder cancer. The third publication was based on a retrospective analysis of molecular test results conducted on a group of patients suspected of having hereditary cancer predispositions. The study presents the frequency of detection of specific variants in the studied population and discusses the advantages of next-generation sequencing in certain clinical situations. The genetic research applications presented in this dissertation exemplify their effective use as diagnostic tools in oncology, enabling precise tumor identification and molecular classification, as well as the identification of hereditary cancer predisposition syndromes.