EP003 Organization of screening for prostate cancer in carriers of germinal mutations in the BRCA1 / 2 genes

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Introduction & Objectives: Hereditary malignant neoplasm occupy a special position, due to young-age development, the increased risk of metastasis, as well as poor prognosis. As known, relatives of patients with mutations in the BRCA 1/2 genes has increasing risk of developing prostate cancer. Studying the risk factors and the duration of the screening will allow us to determine the development of prostate cancer in the early stages. The aim of our study is organization of a screening study to identify patients with germinal mutations in the BRCA1 / 2 genes leading to the development of prostate cancer.

Materials & Methods: This study is based on the analysis of results of molecular genetic testing of blood by sequencing of the “new generation” NGS.

Results: 119 patients with breast and ovarian cancer, mutations in the BRCA 1/2 genes were detected.105 patients has pathogenic mutations in the BRCA1 gene, and 14 probands has mutations in the BRCA 2 gene. After determining the mutations, all probands patient was consulted by a geneticist. A genealogy was compiled; relatives were identified, probable carriers of mutations. Relatives of patients are informed about the need for molecular genetic analysis to identify specific mutations. The study group consists of relatives of patients who are supposed to have a genetic predisposition to cancer. Sanger sequencing determines the specific pathogenic mutations characteristic of the identified patient mutations. When an appropriate mutation is identified, a set of measures is organized for the prevention and early diagnosis of the development of malignant neoplasms.

Conclusions: 119 patients with breast and ovarian cancer, mutations in the BRCA 1/2 genes were detected.105 patients has pathogenic mutations in the BRCA1 gene, and 14 probands has mutations in the BRCA 2 gene. After determining the mutations, all probands patient was consulted by a geneticist. A genealogy was compiled; relatives were identified, probable carriers of mutations. Relatives of patients are informed about the need for molecular genetic analysis to identify specific mutations. The study group consists of relatives of patients who are supposed to have a genetic predisposition to cancer. Sanger sequencing determines the specific pathogenic mutations characteristic of the identified patient mutations. When an appropriate mutation is identified, a set of measures is organized for the prevention and early diagnosis of the development of malignant neoplasms.