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Screening of the pathogenic variant c.3751dupA of the *BRCA2* gene in women from the Republic of Bashkortostan

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Motivation and Aim: Ovarian cancer (OC) is one of the most common malignant neoplasms, the incidence of which has remained consistently high over the past decades. According to the World Health Organization (WHO), in 2020, 313,959 new cases of ovarian cancer were registered in the world, and the number of deaths from this pathology was 207,252 people [1]. According to Kaprin et al. (2021), more than 13,000 cases of this pathology are diagnosed annually in Russia, about 8,000 of which are fatal [2]. One of the important risk factors for OC is hereditary predisposition. The high risk of this pathology is primarily associated with mutations in the tumor suppressor genes *BRCA1* and *BRCA2.* As a result of previous targeted sequencing of DNA samples from patients with hereditary ovarian cancer in exon 11 of the *BRCA2* gene, we identified the pathogenic variant c.3751dupA, which leads to a frameshift (p.Thr1251AsnfsTer14).
The aim of this work was to screen the c.3751dupA variant of the *BRCA2* gene in a group of OC patients and individuals in the control group. *Materials and Methods*: DNA samples isolated from venous blood of patients with hereditary forms of OC (n = 70), sporadic ovarian cancer (n = 167) and women without cancer at the time of blood sampling (n = 322) from the Republic of Bashkortostan were used as research material. Genotyping was performed by high resolution melting curve analysis (HRM).
Results: In our study among patients with OC and the control group, the c.3751dupA/ *BRCA2* variant was identified in one patient with a diagnosis of hereditary ovarian cancer (0.01 %). By ethnicity, the carrier of the studied variant is a Tatar. The manifestation of the disease occurred in postmenopausal women. The carrier of the studied variant by ethnicity is a Tatar. A patient with this pathogenic variant was diagnosed with stage III adenocarcinoma. The tumor was detected in the postmenopausal period.
Conclusion: Thus, our data indicate a low frequency of occurrence of the c.3751dupA/ *BRCA2* variant in women from the Republic of Bashkortostan.

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