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The reduction of two *BRCA1* gene mutations frequencies in ovarian cancer patients from Ukraine

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ABSTRACT

Background: In Ukraine, 3539 patients were diagnosed with ovarian cancer in 2018, and more than half of these cases were lethal. One of the risk factors for ovarian cancer development are the alterations in the breast cancer gene 1 *(BRCA1)*. Among these alterations, the most frequent is the germline mutation 185delAG and the frameshift mutation 4153delA. The frequencies of these mutations were identified earlier (Tsip et al., 2019) in ovarian cancer patients from Ukraine.
Objective: The present study aimed to evaluate the frequency of the mutations 185delAG and 4153delA in the *BRCA1* gene in an unselected ovarian cancer patient's cohort from Ukraine.
Materials and methods: We screened 663 ovarian cancer patients diagnosed at different ages by routine allele-specific polymerase chain reaction (PCR) and real-time PCR.
Results: 13 cases of two *BRCA1* mutations were detected (4 cases of 185delAG and 9 cases of 4153delA).
Conclusions: Out of 663 ovarian cancer patients, 4 cases with 185delAG and 9 cases of 4153delA in the *BRCA1* gene were identified, giving a frequency of 2.0 ± 0.5%. Our data shows that the previous mutations screening results in ovarian cancer patients from Ukraine were higher than in the present study.

1. Introduction

The next-generation sequencing technology opened a new era in cancer research through rapidly increasing the amount of newly generated data and through substantially reducing their costs. Hence, the ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Consortium disclosed the analyses of 2658 whole-cancer genomes and their matching normal tissues across 38 tumour entities. In most cases, when combining coding and non-coding components, cancer genomes contained 4–5 driver mutations which promoted carcinogenesis (Campbell et al., 2020). Another paper published by the same Consortium confirmed a significant role of *BRCA1* germline mutations in the development of breast and ovarian cancers (Li et al., 2020). For this purpose, scientists from 750 institutions across four continents joined their efforts and finances. Is that a lot or a little? From the point of understanding the fundamental mechanisms underlying cancer development, it is a lot. However, the implication of the whole-genome sequencing results for improving treatment is still ineffective since this

Abbreviations: BRCA1, Breast cancer gene 1; CFR, case fatality rate; PCR, polymerase chain reaction; SARS-CoV-2, severe acute respiratory syndrome coronavirus 2.

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