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

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Introduction

- Ovarian cancer (OC) is one of the most common malignant neoplasms, the incidence of which has remained consistently high over the past decades. The high risk of this pathology is primarily associated with mutations in the tumor suppressor genes BRCA1 and BRCA2.

The aim

- 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.

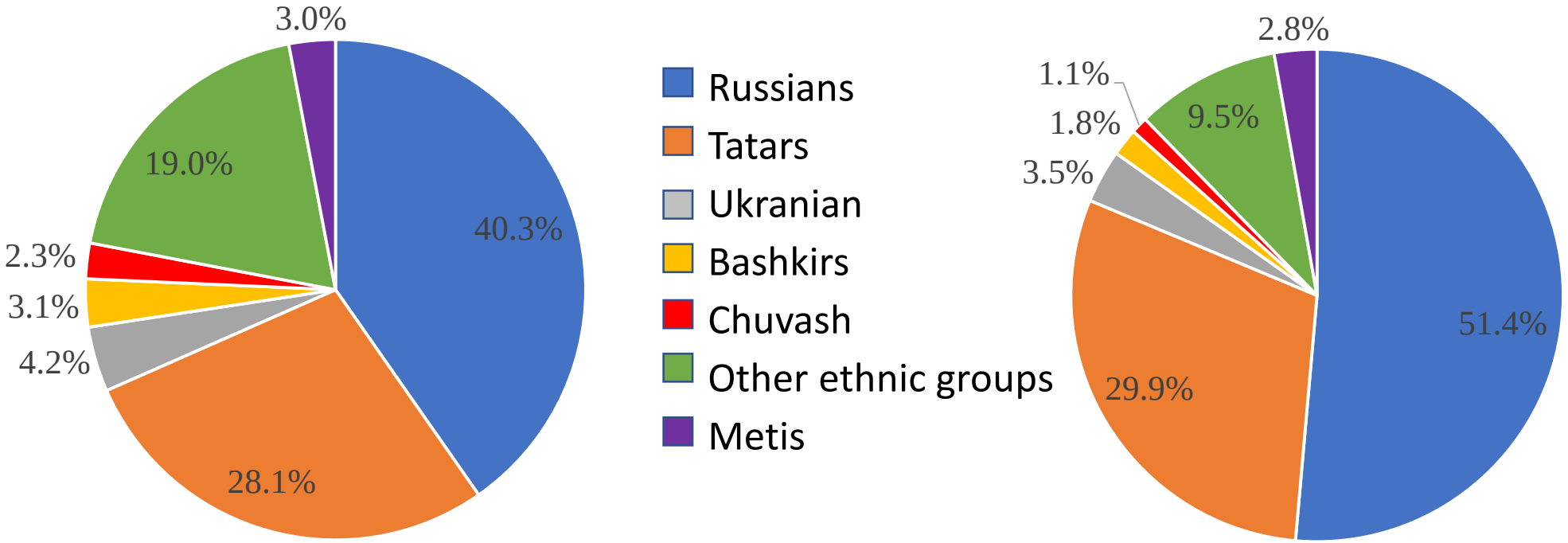
Materials and methods

DNA samples

- Patients with OC (n=332): sporadic OC (n=226) hereditary OC (n=85)
- Women without cancer (n=332)

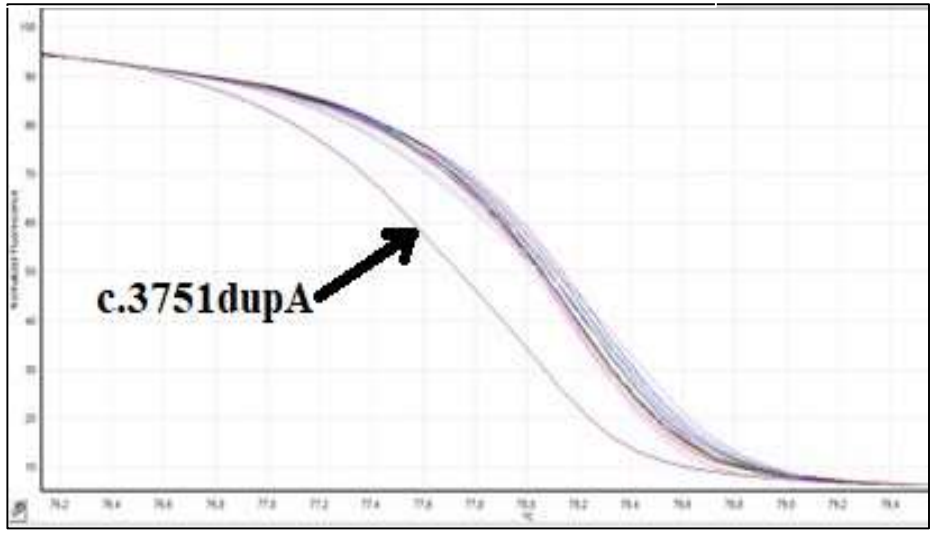
Methods

- Isolation of DNA from peripheral blood by phenol-chloroform extraction
- High-Resolution Melting Curve Analysis (HRM)

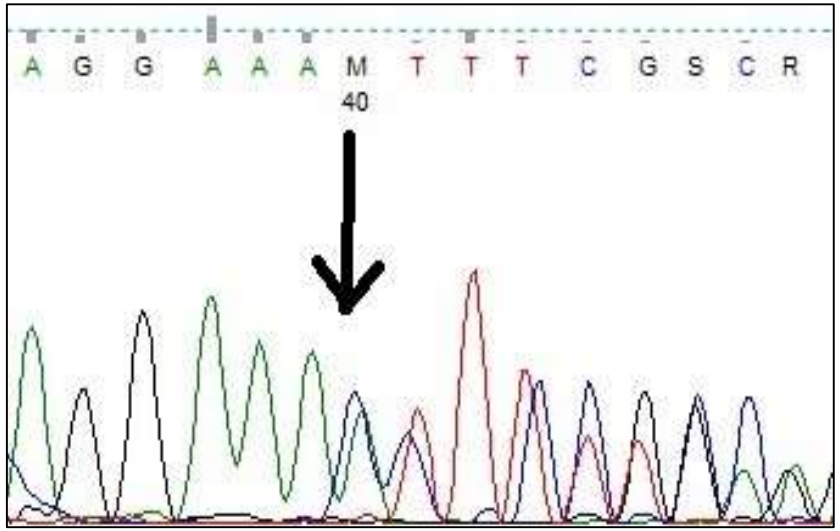


Results

a)



b)



a) Normalized plot of HRM curves for samples without mutation and with mutation c.3751dupA/BRCA2

b) Fragment of the nucleotide sequence of the BRCA2 gene with the c.3751dupA mutation

Conclusion: The obtained results indicate a low frequency of occurrence of the c.3751dupA/BRCA2 variant among OC patients from the Republic of Bashkortostan.

We have found one carrier of the c.3751dupA/BRCA2 variant among patients with hereditary ovarian cancer in a heterozygous state

Clinical Data

-Tatar by ethnic origin
-Adenocarcinoma at 3 stages of the tumor process
- OC was diagnosed in the postmenopausal period

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