

Local features of germinal mutations incidence in cancer patients in Republic of Bashkortostan.

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Abstract:

Presence of germinal mutations in cancer patients plays an important role in expansion of malignant neoplasms. Frequency of different germinal mutations in a population of cancer patients helps to conduct prophylactic measures as in healthy population and in cancer patients.

The aim of this work is to determine region-specific germinal mutations in cancer patients.

Materials and methods:

Study base: Republican Clinical Oncology Dispensary - Ufa, Ufa, Russian Federation

Study period: 2018-2021

Region: Republic of Bashkortostan.

Population:

- patients with manifestation of malignant neoplasm at a young age
- men with burdened family history (cancer incidence in every generation, young age of cancer manifestation)

Investigated cancer types: breast cancer, ovarian cancer, prostate cancer, pancreatic cancer.

Testing method: “next-generation” sequencing method (NGS).

Next mutation types were revealed as region-specific:

BRCA 1: c.5266dupC, c.3143delG, c.5161C>T, c.5382 insC, c.3819delGTAAA, c.300T>G, c.5136G>A, 185delAG, 4153delA, 2080delA;

BRCA 2: c.6621_6622del, c.39-1_39delGA, c.961_962insAA;

CHEK2: c.470T>C, c.444+1G>A;

PALB2: c.1592delT;

RAD50: c.2157delA;

MLH1: c.1637A>G;

MSH2: c.815C>T,

MSH6: c.2554_2556del;

STK11: c.368A>G.

