Identification of gene mutations in patients with breast cancer in a region located in the southeast of the European part of Russia

Poster Nb 25P

ABSTRACT:

It is known that 5-10% of all breast cancer cases have a hereditary component[1,2], of which 20-50% of cases are explained by germline mutations in the genes for susceptibility to breast cancer with high penetrance, the most famous of which are BRCA1 and BRCA2, which are responsible for predisposition to ovarian cancer and breast cancer. The information obtained about the driver's mutations of patients makes it possible to assess both the epidemiological situation in the population and the genetic risk of developing a malignant neoplasm of the proband's relatives who applied for consultation. Correct collection and analysis of the pedigree with the subsequent identification of mutations and genetic counseling makes it possible to provide timely measures for the diagnosis of the risk of developing cancer and its prevention in families with a hereditary predisposition to cancer.

GOALS:

Assessment of the spectrum and frequency of mutations in the genes of hereditary breast cancer in patients from Republic of Bashkortostan

MATERIALS and METHODS:

-100 unrelated patients from republic of Bashkortostan with breast cancer and anamnestic signs of hereditary cancer (1 or more).

-NGS (next-generation sequencing) for tumor tissue material fixed in formalin

PATIENTS CHARACTERISTICS:

Ranges of age: 26 to 73 years

Average age:49,9± 0,94 years

HISTOLOGY TYPE:

Invasive ductal carcinoma – 75%

Invasive lobular carcinoma – 23%

Medullary carcinoma – 2 %

BILOGICAL SUBTYPES:

Luminal A – 28%

Luminal B – 14%

Luminal B Her2positive – 24%

Triple-negative – 28%

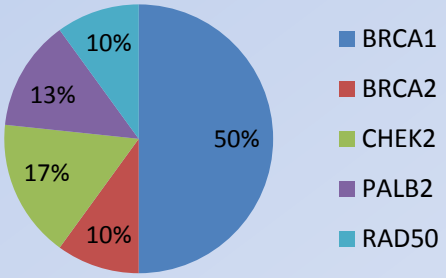
Her2positive – 6%

INCLUSION CRITERIA FOR ONCOLOGY HEREDITERITY:

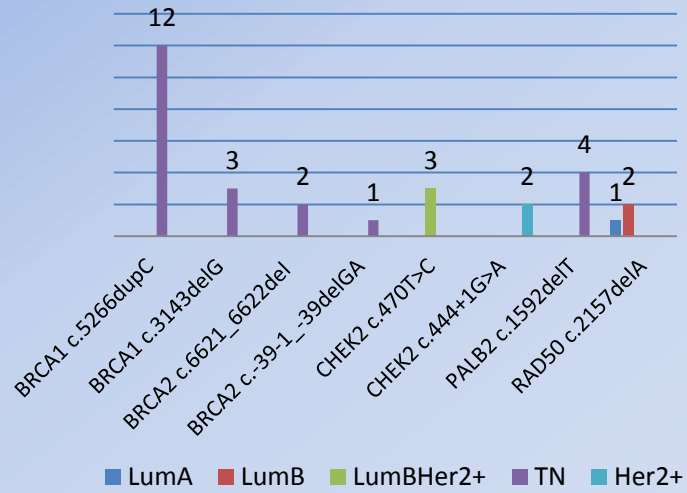
- at least 1 blood relative of the 1st degree of relatives with oncology disease
- multiple tumors cases
- triple-negative cancer in patients under 40 years

RESULTS:

30 of 100 patients had a pathological mutation



MUTATION TYPES AND BIOLOGICAL TYPES OF CANCER



Average age at diagnose:

Mutation found - 42,7±2,77 years

Mutation not found - 52,8 ± 3,82 years

CONCLUSION:

A next-generation sequencing method has significantly improved the efficiency of detecting mutations in the genes responsible for hereditary breast cancer. Pathogenic mutations in the BRCA1 / 2, CHEK2, PALB2, RAD50 genes were found of patients with a hereditary feature of the disease (proband has 1 to 3 blood relatives with malignant neoplasms). The identification of highly penetrant mutations in probands allowed us to determine their relatives, the expectable carriers of mutations, which were informed of the need genetic counseling.

AUTHORS:

Sultanbaev AV (rkodrb@yandex.ru),

Nasretdinov AF, Menshikov KV, Musin SI, Sultanbaeva NI

Republican Clinical Dispensary of Oncology, Ufa, Russia

1. Bochkova N.P., Ginter E.K., Puzireva E.P. Nasledstvennye bolezni. Natsional'noe rukovodstvo. (Inherited Diseases. National Guidelines.) M.:GEOTAR-Media. 2013; 936 p.

2. Onkologiya (Oncology). D. Kaschiato ed. M.:Praktika. 2008; 1039.