PP-BSTR-12

Prevalence of *BRCA1* and *BRCA2* Mutations in Breast Cancer Patients in Russia

A POLONIKOV[1], I ARISTOVA[2], W LEITANTHEM[2], T APPUSAMY[2], A JAISWAL[2], A DOLZHIKOV[2], N PAVLOVA[2], O LUGOVAYA[3], M CHURNOSOV[2]
[1]Kursk State Medical University, Russia, [2]Belgorod State University, Russia

Background & Hypothesis:
Breast cancer (BRCA) is the most common neoplastic disease in women. Being mostly sporadic, about 5-10% of cases are genetically determined. Women with inherited mutation in the BRCA genes have a significant increase in risk of breast cancer and certain other cancers. *5382insC, 185delAG, 4153delA* in *BRCA1* and *6174delT* in *BRCA2* genes are the most known mutations in these tumour suppressor genes.

Methods:
The study enrolled 217 women (mean age 54.2 ± 7.5 years) with breast cancer from the regional oncology dispensary with histologically verified diagnosis and 160 healthy women (mean age 54.4 ± 11.0 years) as a control group. After extraction of DNA from peripheral blood using phenol-chloroform method, all samples were genotyped for *BRCA1* and *BRCA2* polymorphisms by real-time PCR using TaqMan technology for allele discrimination.

Results:
The analysis of the frequency *BRCA1/2* mutations showed that among 217 patients with breast cancer, 7 women had *BRCA1 5382insC* variant (3.2%) and no one had *185delAG, 4153delA* in *BRCA1* and *6174delT* mutations in *BRCA2.* Screenings for these mutations in 160 healthy women showed the homozygosity status.

Discussion & Conclusion:
Compared with some other published papers, this one confirms frequent occurrence of *BRCA1 5382insC* variant in geographically distant regions of Russia, such as Moscow (3.5%), St Petersburg (4.0%), and Siberia region (1.92%). The presented data point to a conspicuous contribution of *BRCA1 5382insC* mutation in breast cancer development in Belgorod region of Russia that may justify an extended *BRCA1 5382insC* screening among women of our population.