Frequency of sequence variants BRCA1, BRCA2 and candidate genes in the Czech Republic

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Objectives: Breast cancer (BC) is one of the most discussed topics and it´s the most common cancers in women. BC are diagnosed over 6500 new cases for year in the Czech Republic. Risk factors include hormonal, nutritional, environmental and genetic factors. Genes play an important role in BC diagnosis are BRCA1/2 genes. In the Czech statistics, the lifetime cumulative risk of BC with confirmed mutations in BRCA1/2 genes ranges from 55 to 85%. The aim was to determine the presence of a pathogenic sequence variant in the BRCA1/BRCA2 genes, the frequency of mutation and finding the percentage of sick women and presence of mutation based on molecular genetic analysis.

Methods: 2033 probands were genetically tested for the presence of sequence variants in BRCA1/BRCA2 genes. Of these, 157 women already had BC, 26 women had ovarian cancer and 22 probands was healthy at the time.

Results: From genetically tested probands were confirmed 199 mutations in BRCA1/2 genes and 13 mutations in candidate genes. This confirms 9.8% of high-risk persons or already cancer patients are hereditary by mutation in the BRCA1/BRCA2 gene. The BRCA1/2 gene mutations were compared in the Czech population and a higher quantity was find in the BRCA1 gene variants. The most frequent sequence variant is c.5266dupC in the BRCA1 gene.

Conclusion: The results show 72% of women with confirmed BRCA1 mutation had BC and 16.4% had ovarian cancer. And women with confirmed mutations in the BRCA2 gene, 77.5% had breast cancer and 7% had ovarian cancer. Compared to Western European countries, the result is similar. The frequency of mutations in the BRCA1/BRCA2 genes is higher in the Czech Republic than the Eastern continent, as is the quantity of female BC patients. We should thing the causes and needs of investigating women with BC predisposition.

Biography

Petra Matulová is a PhD student in Public Health at the Department of Epidemiology and Public Health, Faculty of Medicine, Ostrava. She studies the prevention of individual diseases and also deals with the early diagnosis of cancer. She worked in the Laboratory of Molecular Biology, Department of Medical Genetics and now she is a member of the team at the Center of Epidemiology Research, Faculty of Medicine, Ostrava. She finds that genetics play an increasingly important role in the development and progression of various cancers. She deals with the issue of cancer too.

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