

# **RELATIVELY HIGH INCIDENCE OF NON-FOUNDER BRCA1/2 MUTATION CARRIERS AMONG FAMILIAL BREAST CANCER CASES IN LATVIA.**

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## Abstract

### Background

In Latvia, pathogenic BRCA1 founder variants contribute to about 3.77% of all consecutive primary breast cancers and about 9.9% of all consecutive primary ovarian cancers. Identifying germline pathogenic gene variants in patients with primary breast and ovarian cancer could significantly impact the patients' medical management.

### Methods

10 female and 1 male patients were included. 9 probands had breast cancer, 1 – ovarian and 1 breast and ovarian cancer. In 26 gene panel following genes were included - ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, FAM175A, MEN1, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53, XRCC2. All patients were tested negative for the pathogenic variants of the BRCA1 with founder effect (c.181T>G, c.4035delA, c.5266dupC).

### Results

Of the 11 patients tested, pathogenic variants were identified in 7 (64%) patients: 6 patients carried pathogenic variants of the BRCA1 gene and one of the BRCA2 gene. In 3 patients a variants of uncertain significance of the BRCA2, RAD50 and MRE11A genes was found.

### Conclusion

A 26-gene panel second-line testing increased the number of positive test results in unsolved primary breast and ovarian cancer patients matching criteria for BRCA1/2 testing.