



MEETING ABSTRACT

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Large genomic rearrangements in BRCA1 and BRCA2 genes in breast and ovarian cancer families in Poland

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Mutations in the BRCA1 and BRCA2 genes predispose women to breast and ovarian cancer. The large majority of the alterations identified in these genes are point mutations and small insertion/deletion. However, an increasing number of large genomic rearrangements (LGRs) are being identified, especially in BRCA1. To date just a few large genomic rearrangements of BRCA1 gene have been reported in Poland. Technical limitations of conventional PCR-based methods are cause that gross rearrangements can be overlooked. It has been suggested that about 30% of mutations in the BRCA1 gene are missed by standard mutation detection methods. We screened for LGRs in BRCA1 and BRCA2 genes by Multiplex Ligation-dependent Probe Amplification (MLPA) in 200 unrelated patients with strong family history of breast and/or ovarian cancer negative for BRCA1 Polish founder mutation. We identified 3 different LGRs in BRCA1 gene. No large LGRs were detected in BRCA2 genes.

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