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# ANALYSIS OF THEIR DELETIONS AND DUPLICATIONS IN THE *BRCA1* AND *BRCA2* GENES IN PATIENTS FROM GOIÁS-BRAZIL WITH SUSPECTED OF HEREDITARY BREAST AND OVARIAN CANCER SYNDROME

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Breast cancer is the second most common cancer in the world, and the most common among women population, about 5% to 10% of cases are hereditary and half of them are caused by hereditary breast and ovarian cancer (HBOC), caused by variations in *BRCA1* and *BRCA2* genes. The present study aimed to identify deletions and duplications prevalence in *BRCA1* and *BRCA2* genes on breast cancer patients in Goiás, Brazil, and was approved by the National Research Ethics Committee according with number of CAAE 50626315.6.0000.5078. Were evaluated 46 breast cancer's patients who fulfilled National Comprehensive Cancer Network (NCCN) criteria for HBOC syndrome screening. About 4 mL blood sample was collected from each patient for DNA extraction using commercial kit and the Multiplex Ligation Dependent Probe Amplification (MLPA) technique was performed using the SALSA MLPA P002 *BRCA1* and SALSA MLPA P045 *BRCA2/CHECK2* kits. The majority of the patients were female (97.83%) and the mean age of the patients was 37.52 years. In this group, 43.47% of the patients were younger than 35 years at the time of diagnosis and 35% of them were diagnosed with triple negative tumors. The most common molecular subtype was luminal A (46.2%) followed by triple negative tumors (28.20%). Four family history profiles were observed, which 45.65% had a family history of breast cancer and other cancers, 33% had a family history of cancer without cases of breast cancer, 17.39% had only a personal history of cancer in the family and 4.34% of the patients had only a family history of breast cancer. No patient was found with rearrangements in the *BRCA1* gene. In the *BRCA2* gene, one patient (2.12%) presented a false positive result for the heterozygous deletion of the 27 exon, which may have been caused by the presence of a small change in the probe binding region. This was the first study performed to analyze large deletions and duplications in patients from the central-western region of Brazil. Can be conclude that the frequency of large deletions and duplications in the *BRCA1* and *BRCA2* genes is low in the population of Goiás.
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