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## BRCA1 AND BRCA2 germline pathogenic variants in Brazilian breast cancer patients from a private oncologic service in Goiânia, Goiás

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**Objective:** The objective of this study was to evaluate the clinical and pathological characteristics of a group of breast cancer (BC) patients presenting germline pathogenic variants (PV) in the BRCA1 and BRCA2 genes. **Methodology:** This descriptive study was approved by the Research Ethics Committee of PUC Goiás and comprised the retrospective analysis (clinical, genetic, and histopathological) of patients with BC and PV in BRCA1/BRCA2 genes, treated at a tertiary oncologic service. **Results:** Among 52 patients, 51 were women. The average age for the group was 42.3 years ( $\pm 11.8$  years). An expanded germline genetic panel was performed in 88.6% of the cases, resulting in 29 BRCA1 PV (55.8%) and 23 BRCA2 PV (44.2%). The median time between BC diagnosis and genetic testing was 3 months. Genetic testing was performed before surgery in 72.0% of the patients with a BRCA1 PV and in 39.0% of those with a BRCA2 PV. The histological type was carcinoma without other specifications in 86.5% of cases and 48% presented histological grade 3. Among BRCA1 PV, estrogen receptors (ER) were negative in 79% and ER >10% in 78.3% among BRCA2 PV. Cancer in the contralateral breast was detected in 15.4% of the patients. Prophylactic contralateral mastectomy and salpingo-oophorectomy were performed in 73.0% and 57.7% of patients, respectively. With a follow-up period of at least 3 years in 78.8% of the patients, 92.3% were alive and four deaths were recorded, all of which occurred in patients with a BRCA2 PV, and of these, three evolved with progressive disease in the central nervous system. **Conclusion:** This is the largest retrospective epidemiological study including BC with BRCA1 and BRCA2 PV in the Central-West region in Brazil and one of the largest institutional series of BC patients with BRCA1 and BRCA2 PV in the country. With public policies that improve access to genetic testing in Goiás, new data are expected.

**Keywords:** gene BRCA1; gene BRCA2; genetic variant; mutation; breast neoplasm.