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Genetic screening of pathogenic variants in relatives of patients with hereditary breast and ovarian cancer in the state of Goiás: tool for early diagnosis and prevention of breast and ovarian cancer in the Goiás Todo Rosa Program

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Introduction: Approximately 10% of breast and ovarian cancer cases are hereditary. Genetic screening of family members of patients with germline pathogenic variants is essential for prevention, early diagnosis, and personalized therapeutic actions. Identifying pathogenic variants allows the screening of family members at risk. This strategy, already incorporated into the Unified Health System (SUS) in Goiás, strengthens public policies and enables prevention and precision oncological care. **Objective:** This study aimed to identify germline pathogenic variants and their prevalence in family members of patients with pathogenic variants. **Methods:** A total of 308 patients who met the National Comprehensive Cancer Network criteria for suspected hereditary breast and ovarian cancer syndromes were evaluated and referred to the Human Genetics Center of the Federal University of Goiás by reference hospitals of the SUS in the state of Goiás. After pre-genetic counseling and application of the free and informed consent form, 4 mL of venous blood were collected for deoxyribonucleic acid (DNA) extraction used for next-generation sequencing with the Oncomine™ BRCA Expanded panel kit and submitted to sequencing on the Ion Torrent platform. **Results:** Of the 308 patients evaluated, 5.8% (18/308), 1.8% (6/308), and 3.9% (12/308) were positive for some pathogenic variant in the BRCA1, BRCA2, and TP53 genes, respectively. Three families were investigated for the variant in the TP53 gene and two families with a pathogenic variant in the BRCA1 and 2 genes. The variants c.1010 G>A (6/7) and c.455C>T (1/7) were identified. Within the families with variants, it was observed that 47.1% (8/17) of the tested relatives were also positive cases, with one case of cancer followed by death. **Conclusion:** Given these results, genetic screening of relatives of patients with variants in BRCA1 and 2 genes enables prevention, early diagnosis, and personalized approaches, expanding the impact of the healthcare network in Goiás.

Keywords: breast cancer; Li-Fraumeni syndrome; tumor suppressor protein p53; hereditary breast and ovarian cancer syndrome.