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Genetic testing impact on clinical decision-making in Brazilian breast cancer patients with identified genetic variants

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Objective: The present study aimed to analyze the impact on clinical decision-making of an identified pathogenic or likely pathogenic genetic variant in breast cancer Brazilian patients. **Methods:** This is a retrospective observational cohort of breast cancer patients with known pathogenic variants (PV), likely pathogenic variants (LPV), and variants of uncertain significance in cancer-related genes in four private and one philanthropic institution in Minas Gerais, Brazil. Pathology and medical records were reviewed, and patients were interviewed. **Results:** A total of 62 patients were included. Forty-three patients with breast cancer and a PV/LPV underwent primary breast surgery; 22 of them (51.1%) were aware of the genetic variant at the time of surgery. In the group of patients who were aware of the variant at surgery, 68.2% underwent bilateral surgery; in the group not aware, only 4.7% underwent bilateral mastectomy ($p < 0.001$). The median time from diagnosis to genetic testing was 94.8 months in patients diagnosed before 2015 and 4.8 months in those diagnosed after 2015 ($p < 0.001$). Thirty-eight patients (57.5%) had access to genetic counseling in the entire cohort, and only 3 of 18 (16%) were from the public sector. Nineteen different PV/LPV were found in BRCA1, 13 in BRCA2, six in TP53, four in ATM, and four in PALB2. The most common PV was c.2T>G (p.Met1Arg) in BRCA2, present in four unrelated patients. **Conclusion:** This analysis shows a significant delay in genetic testing and limited access to counseling, which has influenced the high rate of conservative surgery for patients unaware of their genetic diagnosis at the time of surgery. These findings underscore the urgent need to expand hereditary cancer testing and counseling services in Brazil.

Keywords: breast neoplasms; hereditary breast and ovarian cancer syndrome; BRCA1 gene; BRCA2 gene; prophylactic mastectomy.