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# 1028 – Germline mutations and clinical outcomes in hereditary breast cancer: a retrospective cohort from northeastern Brazil

Samuel Castro Campos<sup>1</sup>, Sabas Carlos Vieira<sup>2</sup>

<sup>1</sup>Universidade Federal do Piauí – Teresina (PI), Brazil.

<sup>2</sup>Oncocenter – Teresina (PI), Brazil.

**Introduction:** Germline mutations in breast cancer susceptibility genes are associated with aggressive tumor biology and specific therapeutic vulnerabilities. Epidemiological data on mutation carriers diagnosed with breast cancer in northeastern Brazil remain scarce.

**Methods:** A retrospective cohort of 42 women with germline pathogenic or likely pathogenic variants and breast cancer, treated at Oncocenter, Teresina, Brazil. Records were reviewed between November 2025 and March 2026. Clinical, genetic, pathological, treatment, and outcome variables were analyzed descriptively. This study was approved by the Research Ethics Committee of the State University of Piauí (UESPI) under CAAE 30154720.0.0000.5209.

**Results:** Median age at diagnosis was 40.5 years; half of the patients were aged 40 years or younger. Family history of breast cancer was present in 92.9%. BRCA1 was the most frequently mutated gene (50%), followed by BRCA2 (26.2%); 90.5% of variants were classified as pathogenic. The predominant molecular subtype was triple negative breast cancer (TNBC; 51.4%). Neoadjuvant chemotherapy was administered in 75% of the cases; complete pathological response (pCR) was achieved in 44.4% of evaluable patients. Poly (ADP-ribose) polymerase (PARP) inhibitors were used in 20.6% of patients, predominantly olaparib in the adjuvant setting. At final follow-up, 87.8% were alive and disease-free; disease-related mortality was 7.3%.

**Conclusion:** The cohort shows a predominance of BRCA1 mutations and triple negative tumors in young women, consistent with the known biology of hereditary breast cancer. The 44.4% pCR rate reflects platinum sensitivity associated with homologous recombination deficiency. PARP inhibitor use was low, pointing to underutilization in this regional setting. These data characterize hereditary breast cancer in a population with limited published data from northeastern Brazil.

**Ethics:** Research Ethics Committee Approval (CAAE 30154720.0.0000.5209).

**Keywords:** breast neoplasms; neoadjuvant therapy; germ-line mutation.