https://doi.org/10.29289/259453942025V35S1129

Genetics and predisposition to breast cancer: the impact of mutations in the BRCA1 and BRCA2 genes

Camila Carvalho Lenti[1], Amarílis de Oliveira Almeida[2], Isabella Siciliano Montalto[3], Luana Camin Oliveira[2], Marina Barros Dotto[4], Pedro Henrique Veloso de Souza[3]

[1]Faculdade de Minas Belo Horizonte – Belo Horizonte (MG), Brazil.
[2]Universidade Nove de Julho – São Paulo (SP), Brazil.
[3]Universidade Municipal de São Caetano do Sul – São Caetano do Sul (SP), Brazil.
[4]Universidade Santo Amaro – Santo Amaro (SP), Brazil.

Introduction: Breast neoplasia is influenced by environmental, hormonal, and genetic factors. Among the hereditary factors, mutations in the BRCA1 and BRCA2 genes stand out, as they are strongly associated with disease predisposition, with a risk of 80–85% for the development of breast cancer when present. **Objective:** The aim of this article was to analyze the impact of mutations in the BRCA1 and BRCA2 genes on breast cancer predisposition, highlighting their influence on the risk of the disease and the screening and prevention strategies available. **Methods:** This is a descriptive epidemiological study based on data from the Cancer Information System (SISCAN) referring to the risk elevated by screening mammography in Brazilian residents between 2019 and 2025. **Results:** Genetic factors play a key role, especially BRCA1 and BRCA2 mutations, which increase lifetime risk by 50%. Rigorous follow-up on genetic testing enables preventive strategies like prophylactic mastectomies. Early detection through mammograms remains crucial for reducing mortality, and for this purpose, mammography is recommended from the age of 50. A public policy initiative from the 1980s has improved access to screenings and early detection rates. The SISCAN analysis evaluated 2,130,098 high-risk breast cancer patients, 726,812 with a family history. Mammography adherence was high, with only four patients not undergoing the exam. Breast Imaging Reporting and Data System (BI-RADS®) category 5 was found in 1,842 cases and category 4 in 8,054. The study suggests a stricter screening, including magnetic resonance imaging and genetic testing, to improve early detection. **Conclusion:** The analysis of SISCAN data demonstrates the importance of screening for breast cancer. This tool remains ideal for early detection, resulting in positive data to reduce mortality, although there are still challenges such as inequality in access to health. Public policies have proven effective in expanding coverage, due to the inclusion of magnetic resonance imaging and genetic tests.

Keywords: breast cancer; mammography; genetics.