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Epidemiological and histo-molecular profile of patients with breast cancer who underwent genetic testing at a tertiary clinic in northeastern Brazil

Débora Medeiros de Carvalho¹, Kamila Bezerra Fernandes Diocesano², Sabas Carlos Vieira³, Maysa Gabriela Costa Cruz⁴, Fernanda Jorge Martins⁴, Camila Holanda de Sousa⁴, Nívyá Emanuele Vilarinda dos Santos⁴

¹Centro Universitário UniFacid – Teresina (PI), Brazil.

²Universidade Federal de Piauí – Teresina (PI), Brazil.

³Oncocenter – Teresina (PI), Brazil.

⁴Centro Universitário Uninovafapi – Teresina (PI), Brazil.

Objective: The aim of this study was to describe the epidemiological and histo-molecular profile of patients with breast cancer (CAM) who underwent genetic testing at an oncology clinic in northeastern Brazil. **Methodology:** This is a retrospective cohort study on patients with CAM who underwent genetic testing from 1998 to 2022 at a tertiary clinic in northeastern Brazil and who underwent multigene panel testing for hereditary cancer predisposition syndromes. **Results:** Data were collected from 208 patients, of which 122 had CAM. Of these, 14.75% were BRCA1, 11.47% were BRCA 2, 15.57% were VUS, 4.09% were other high penetrance mutations, and 54.12% did not have mutations. The most prevalent histological type was non-special carcinoma (42.62%) and the second was carcinoma in situ (37.70), 3.27% micropapillary, 5.73% lobular, and 0.81% inflammatory. The most prevalent molecular type in the sample was HER 2 (44.26%), and triple-negative was the second most prevalent (16.39%), 40% corresponding to luminal HER 2 and luminal. However, among the BRCA 1 and 2 mutations, the most prevalent molecular type was the triple-negative (34.37% of a total of 32 BRCA 1 and 2). The age with the most prevalent CAM was the range of 35–45 years with 32.78%. In addition, 99% of the patients were female and 1% were male (only one male), 43.4% of the tumors were grade 2, and 55.73% of the lymph nodes were not involved, with 17.2% of the only 1 affected lymph node. **Conclusion:** It is concluded that the most significant mutations are in the BRCA 1 and 2 genes with the triple-negative molecular type being the most prevalent in these genes, showing that the results corroborate the data already existing in the literature, as well as the importance of the genetic panel for the best individualization and optimization of treatments.

Keywords: breast cancer; genetic testing; epidemiology.