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HEREDITARY BREAST CANCER IN THE PUBLIC HEALTH SYSTEM OF FEDERAL DISTRICT (DF) – BRAZIL

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Objective: The lack of financial resources challenges the inclusion of genetic testing in the Brazilian Public Health System. This study aims to describe the detection rate of germline pathogenic variants (GPVs) in patients at risk of hereditary breast cancer (BC) in the public hospitals of Brasilia, DF, as well as the clinical and demographic profile of patients (pts). **Methods:** Hereditary cancer risk assessment based on the National Comprehensive Cancer Network Criteria, version 1.2020 was performed on patients with a personal history of BC who were being followed in a public hospital (DF) between January 2021 and January 2022. **Results:** Among 217 female pts eligible for this study, 78 pts performed germline multigene panel testing out of pocket. Panels included 26–84 cancer susceptibility genes. Patients in this cohort were mainly from the center-west (46%) and northeast (31%) of Brazil. The median age of BC diagnosis was 42 years. Invasive ductal carcinoma represented 88% of the tumors. From a total of 78 BC, 52% were hormone receptor-positive, 23% HER2 positive, and 24% triple-negative. Most patients presented with locally advanced disease: 50% (n=39) IIB-IIIC and 8% (n=6) had metastatic disease. The detection rate of GPVs was 20% (n=16). Among these 16 patients, the most frequently mutated genes were BRCA1/2 (n=11, 68.5%) and TP53 (n=2, 12.5%). **Conclusion:** The overall detection rate of GPVs was similar to other worldwide studies. In comparison with other Brazilian studies, GPVs in TP53 were at lower rates, possibly because this cohort was enriched by patients from Brazilian center-west and northeast. Higher rates of advanced disease at BC diagnosis may impact treatment outcomes. The lack of access to genetic testing in the public health system takes away the opportunity for cancer prevention, more effective treatments, and proper family risk assessment.

Keywords: Hereditary breast cancer syndrome. Public health. Germline mutation.