Hereditary cancer syndromes in patients with second primary breast cancer

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Objective: The objective of this study was to evaluate the presence of hereditary cancer syndromes (HCS) in patients with a diagnosis of two primary breast carcinomas and analyze the frequency of pathogenic variants in high- and moderate-penetrate genes. Methodology: This is a retrospective unicentric cohort study on patients with a diagnosis of two primary breast cancers, diagnosed between January 2000 to December 2020, at A.C. Camargo Cancer Center, Brazil. The association between categorical variables was analyzed by the chi-square test or Fisher’s exact test. For survival curves, the Kaplan-Meier method and log-rank test were used to describe the survival curve differences. Results: Medical records of breast cancer patients were reviewed from 2000 to 2020, and a frequency of 600 patients with two primary breast tumors (metachronous or synchronous) was observed. In total, 190 (31.7%) patients performed genetic testing and 35 (5.8%) patients presented a pathogenic or likely-pathogenic germline variant in cancer predisposing genes. Conclusion: Our results revealed a low rate of genetic testing among patients with two primary breast cancers in a cancer center and a frequency of carrier patients lower than expected.

Keywords: breast cancer; hereditary cancer syndromes.