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Two pathogenic variants in a patient with cervical and breast cancer: Case report

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Introduction: The presence of two pathogenic germline variants in hereditary cancer is an uncommon event. We report a case of a Brazilian patient from Teresina, Piauí, who developed breast and cervical carcinoma with pathogenic variants in BRCA2 and MUTYH genes. **Case Report:** A 25-year-old female patient in 2012 underwent a radical hysterectomy with pelvic lymphadenectomy without ovarian preservation for treatment of histologic grade 2 (G2) squamous cell carcinoma (SCC) of the cervix, FIGO stage IB2. Histopathology of the surgical specimen revealed SCC, G2, stromal invasion 16 mm, 4.5 cm in diameter, compromised parametrium, 6 lymph nodes without metastasis, and normal ovaries. She received pelvic radiotherapy and brachytherapy associated with platinum-based chemotherapy. In 2017, she was diagnosed with histologic grade 1 invasive breast carcinoma of no special type in the right breast. Immunohistochemistry revealed that it was a luminal B tumor (estrogen receptor (ER)+ 90%, progesterone receptor (PR) + 80%, human epidermal growth factor (HER2) 1+, Ki-67 40%), stage IA (T1N0M0)). Neoadjuvant chemotherapy with doxorubicin and cyclophosphamide (AC, 4 cycles) followed by paclitaxel (12 cycles) was performed. The patient underwent segmental mastectomy, and sentinel lymph node research and histopathology revealed complete pathological response and negative sentinel lymph node residual cancer burden 0. She had a history of three pregnancies and three deliveries, with no case of neoplasia in the family. In 2023, multigene test for hereditary predisposition to cancer was performed, in which two pathogenic variants were detected being one in BRCA2 gene (c.8725A>T) and the other in MUTYH (c.1187G>A). Currently, there is no evidence of active disease and on schedule for colonoscopy, endoscopy, and bilateral risk-reducing mastectomy. **Conclusion:** In young patients with multiple cancers, a search for pathogenic variants related to hereditary cancer predisposition syndromes should be offered, as in the present case.

Keywords: BRCA2 gene; uterine cervical neoplasms; breast cancer.