https://doi.org/10.29289/259453942022V32S1074

553 - SECOND BREAST CANCER IN A WOMAN WITH GENETIC SYNDROME

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Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disease and is the most common neurocutaneous syndrome. It results from a defect in the gene located on chromosome number 17 that produces the protein neurofibromin, involved in controlling cell growth. Women with NF1 have a higher risk of developing breast and contralateral breast cancers. There is a relationship between high estrogen receptor (ER) and worse survival, which is also affected by the low overall life expectancy of patients with neurofibromatosis. Given that data also suggest that there are genes that interact with the *NF1* gene, particularly in relation to the Breast Cancer gene 1 (*BRCA1*) subset. The interaction of altered expression of the NF1 neurofibromin protein in breast cell lines with upregulation of Ras is not inhibited through the PI3K and Raf/MAPK/ERK pathways. Increased PI3K activity has often been related to poor survival and resistance to hormone treatment in ER-negative breast cancer, while elevated Ras/MAPK/ERK activity has been related to metastasis and poor survival in both ER positive and negative. Mutations and deletions in NF1 are even more prevalent in HER2-amplified breast cancer subtypes and in basal tumor subtypes. In fact, all women with NF1, such as the case reported below, should start screening for breast cancer from the age of 30 and not from the age of 50 as in women not affected by the disease, as well as adequate and early counseling of oncogenetic. MSF, 56 years old, female, with neurofibromatosis was treated for invasive ductal carcinoma (ICD) in the left breast, RH negative in 2006, with mastectomy and axillary emptying, followed by adjuvant chemotherapy and radiotherapy. Menarche at age 17, menopause around age 41, at which time she underwent chemotherapy, was nulliparous, and denied hormone use. She had a negative family history. She was admitted to the Mastology Unit of the HBDF in March 2021 with an ultrasound examination of the right breast on February 19, 2021, BIRADs 4 at the expense of a solid, irregular nodular image and imprecise limits at 12 o'clock, measuring 21×16 mm. On physical examination, nodular lesions (neurofibromas) of varying sizes were observed, distributed throughout the trunk and limbs, and a 3 cm nodulation was palpated in the upper internal quadrante (QSM) of the right breast, close to the NAC with a negative axillae and plastron on the left, staging cT2N0M0 — IIA. Core biopsy confirms CDI, grade II, with ductal carcinoma in situ present, and luminal B-like immunohistochemistry (IHC). Staging tests without an evidence of distant disease. In July 2021, a mastectomy was performed with a sentinel lymph node biopsy (SLNB) on the right in view of the clinical staging and IHC profile, but of the four lymph nodes stained with patent blue, three were positive in intraoperative frozen section biopsy; therefore, the axilla was completed with dissection. The patient was discharged on the first postoperative day with weekly follow-up at an outpatient clinic, and the dressing was discharged in August 2021. Biopsy results confirmed a 6.5-cm ICD, grade III, ICD present with intermediate nuclear grade, and with all disease-free margins. The patient was referred to a clinical oncology but arrived at the oncology more than 120 days after surgery, with time loss for adjuvant treatment.