https://doi.org/10.29289/259453942022V32S1051

467 - LI-FRAUMENI SYNDROME: A CASE REPORT

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Li-Fraumeni syndrome is a disease with an autosomal dominant inheritance of high penetrance and was originally described in 1969. The definitive diagnosis is based on the identification of a pathogenic variant in the *TP53* gene. Birch and Chompret and classical models were used as the clinical criteria to identify individuals who are the candidates for molecular screening. It is responsible for about 1% of hereditary breast cancers and is related to other neoplasms, the most common sarcomas, leukemias, and adrenal carcinoma. Among the differential diagnoses, we can consider pathogenic variants of BRCA1/BRCA2 and Lynch syndrome. The behavior of cancer is usually similar to that of patients without Li-Fraumeni syndrome except for the age of early onset. Screening for the breast cancer with an annual magnetic resonance is recommended in women from the age of 20, colon cancer with colonoscopy every 2 or 5 years from the age of 25, and osteosarcoma and soft-tissue tumor with full-body resonance at an early age. Interventions are recommended for patients with a family history or individuals with a reported mutation. Mastectomy is generally recommended for women with breast cancer due to the risks of a second primary breast cancer or a second radiation-induced cancer. The risk of contralateral breast cancer in patients with TP53 diagnosed under 35 years of age is approximately 4%–7% per year. In this scenario, we bring a case report of a young female patient with synchronous tumors of maxillary osteosarcoma and breast cancer to study the approach, prevention, and guidance in these cases. N.O.B., 21 years old, single, born in São Paulo, nonparturient, mother's death due to breast cancer at age 36, and sister's death due to neuroblastoma at age 2. At the first medical appointment in June 2021, she complained of the presence of a nodule in her left breast persisting for 2 years and reported a palate lesion present for 1 month and with rapid growth, associated with existing oral cavity deformity. On the breast ultrasound examination performed in June 2021, a solid, hypoechogenic, irregular, microlobulated, nonparallel to skin was revealed, measuring 1.5×1.2×1.3 cm at 10 h of the left breast (BI-RADS classification®: 5). The pathological report from a directed biopsy of the nodule showed an invasive carcinoma of nonspecial histological type, estrogen receptor 80%, progesterone receptor 100%, Her2 negative, and Ki67 60% — clinical staging cT1N0. The examination of her palate lesions reported high histological osteosarcoma — cT1N0M0. Considering the double-tumor diagnosis plus the highly aggressive lesion of the patient's palate, the team chooses to start the treatment with partial maxillectomy and bilateral infrastructure surgery, tracheostomy, and reconstruction with a microsurgical flap of the right fibula in July 2021. Later in October 2021, the patient's treatment was followed by a combination of left adenomastectomy, left sentinel lymph node biopsy, and reconstruction with bilateral prosthesis. Due to family and personal history, she went through genetic testing for pathogenic mutation on the *TP53* gene. Fertility preservation was performed with oocyte freezing. As an adjuvant treatment, it is scheduled for four cycles of docetaxel and cyclophosphamide. As prophylactic measures, the patient is expected for an adenectomy in the contralateral breast and to follow up with clinical examinations twice a year, as well as both mammography and MRI once a year.