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Follow-up of a Li-Fraumeni syndrome case

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Introduction: Li-Fraumeni syndrome (LFS) is responsible for about 1% of hereditary breast cancers (BC). We present a case report of a young woman with synchronous osteosarcoma and BC. **Case Report:** NOB, 23 years old. Mother died with BC at 36 years old and sister died due to neuroblastoma at 2 years old. She was referred in 2021 for a nodule in her left breast and the ultrasound results showed an irregular nodule of 1.5×1.2×1.3 cm BI-RADS®5 and anatomopathological invasive carcinoma of non-special histological type (NST), estrogen receptor 80%, progesterone receptor 100%, Her2 negative, and Ki67 60% cT1N0. It was associated with a lesion in the alveolar mucosa with bleeding and deformity of the oral cavity with anatomopathological high histological osteosarcoma-T1N0. Surgical treatment was performed: maxillectomy of meso and bilateral infrastructure+tracheostomy+reconstruction with microsurgical flap of the fibula and, then, left adenomastectomy+sentinel lymph node biopsy+prosthesis reconstruction. Surgical anatomopathological results in central/medullary high-grade conventional osteosarcoma chondroblasts 7.3×6.1×3.9 cm, free surgical margins and four cervical lymph nodes free of neoplastic involvement, and invasive breast carcinoma NST with medullary characteristics 1.8×1.3 cm, free margins, and absence of metastasis in two sentinel lymph nodes –pT1pN0. Genetic test resulted in pathogenic mutation TP53 gene, position chr17:7.674.257, consequence p.Tyr236HisENST00000269305. Adjuvant chemotherapy was docetaxel+cyclophosphamide. Two years after treatment, she is taking tamoxifen, scheduled for contralateral adenomastectomy, and maintains high-risk follow-up. There is no signal of any cancer disease. **Discussion:** LFS is an autosomal dominant inheritance of high penetrance. The diagnosis is based on the identification of a pathogenic variant in the TP53 gene. It is related to several tumors diagnosed at an early age. BC is the most common cancer and affects 27–31% of patients. Osteosarcoma corresponds to 3%–16% of cases, usually occurring before the age of 30 years. The prognosis of patients does not differ from those with sporadic cancer. They must be monitored by a multidisciplinary team, screening with annual whole body/breast MRI and mammography, and colonoscopy every 5 years. Genetic counseling is essential.

Keywords: Li-Fraumeni syndrome; osteosarcoma; breast neoplasms.