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MALE BREAST CANCER ASSOCIATED WITH A LARGE DELETION IN BLM GENE – REPORT OF A CASE

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Malignant breast neoplasm in men is rare, corresponding to less than 1% of all breast neoplasms, and 100 times less frequent than in women. It is molecularly different from female breast cancer, and germline pathogenic mutations in genes aside from BRCAs have been recently associated with an increased risk of male breast cancer. Here, we report an elderly male, 71 years old, with a malignant neoplasm in the left breast, with positive hormone receptors, HER2-negative, and Ki-67 of 25%. A modified radical mastectomy was performed, and the surgical specimen showed a micropapillary invasive mammary carcinoma, 1.9 cm, 2 of 11 lymph nodes positive, pT1cpN1acM0. He was treated with adjuvant chemotherapy and radiotherapy, followed by endocrine therapy. His mother had breast cancer at 50 years, and his smoking father died of lung cancer. During his treatment, a multigene panel was done and a heterozygous likely pathogenic large deletion involving exons 20–22 of the BLM gene was found, associated with a variant of unknown significance in the same gene; c.3427G>A; p.(Glu1143Lys). All his three daughters harbor the same mutation. The risk of breast cancer in association with a heterozygous pathogenic variant in the BLM gene is still controversial because of its ability to cause tumors when not associated with polymorphisms in other homologous recombination genes, which poses a challenge for genetic counseling, surveillance, and management. This report aims to add data and clinical evidence to the attempts to elucidate the role of BLM germline variants in breast cancer predisposition.

*Patient signed an informed consent.

Keywords: Male breast cancer. Cancer predisposition.