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NEUROENDOCRINE CARCINOMA OF THE BREAST AND ILEUM IN A PATIENT WITH BRCA2 PATHOGENIC VARIANT – ONCOLOGIC AND GENETIC CONSIDERATIONS DERIVED FROM A CASE REPORT

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Background: Neuroendocrine neoplasms (NENs) are a heterogeneous group of neoplasms. Most frequently, they occur in the digestive system, and breast neuroendocrine tumors constitute less than 1% of all of them. Germline mutations can increase the risk of developing tumors and predispose to hereditary cancer syndromes. Some NENs are well-established components of common hereditary syndromes. Recently, whole genomic sequencing revealed that 17% of apparently sporadic pancreatic NENs carried germline mutations, including DNA repair genes, such as BRCA2. It is well known that this gene plays a role in hereditary breast cancer, but variations in these genes were not described in patients with breast/ileum neuroendocrine tumors. We present a patient with a neuroendocrine tumor and a germline pathogenic variant in BRCA2. **Case report:** A 44-year-old female patient presented with a palpable lesion at the right breast, with 1.1×1.4 cm, and a biopsy confirmed an invasive ductal carcinoma, grade 2. Immunohistochemistry revealed a neuroendocrine breast carcinoma (ER10% PR5%, HER2 negative, Ki-67=8%). The regular staging examinations did not show any abnormalities, but a 68Ga PET/CT demonstrated an ileum wall thickening with a marked expression of somatostatin receptors compatible with primary disease, with mesenteric lymph nodes and hepatic lesions suggestive of metastasis, in addition to the right breast lesion that could be either a secondary implant or a primary synchronous tumor. A detailed family history did not reveal any important cancer cases in the family except for the father and a paternal uncle, both with prostate cancer at 72 and 85 years old, respectively. Germline genetic analysis confirmed the presence of a heterozygous pathogenic variant in BRCA2 (c.2167delA;p.Ser723Alafs*7). The patient is currently being treated with octreotide LAR with good tolerance and stable disease. **Final comments:** This case shows the importance of molecular germline investigation in patients with NENs. This patient adds knowledge to the association of the BRCA2 gene and neuroendocrine tumors.

Keywords: Breast cancer. BRCA2.