https://doi.org/10.29289/259453942022V32S1019

490 - CONCERNING A FAMILY WITH BRCA2 MUTATION

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Introduction: Breast cancer is the most common malignancy in women and represents a major obstacle to public health worldwide. The molecular diagnosis of this type of cancer is one of the main contemporary challenges in oncology, since it is hampered by a complex inheritance pattern, characterized by both genetic and environmental factors. Only a minority of breast cancers are explained by the presence of high penetrance gene mutations, such as those in the *BRCA1* and *BRCA2* genes, which together with mutations in intermediate penetrance genes explain only up to 25% of the risk. In fact, much of the genetic influence is elucidated by low penetrance variants. Mutations in the germline BRCA1 and BRCA2 are the most common alterations in cases of early onset or of family history of breast cancer. It is also important to acknowledge that BRCA2 mutations can increase the risk of developing other cancers. Some studies show a relation between BRCA2 mutations and the development of leukemia, especially acute myeloid leukemia (AML). Also, some of these mutations, when inherited from both parents, cause a rare form of Fanconi anemia, a syndrome associated with the development of AML. In addition, there are studies evaluating a higher risk of pancreatic and esophageal cancer in carriers of BRCA2 mutations. The risk of colorectal cancer is also increased in patients with BRCA1 mutations. However, there are also some authors who defend that BRCA2 mutations could also be related. The specific statistics are not well defined because of the lack of data focusing on the relationship with the aforecited types of cancers, demonstrating the need for further analysis. This study aims to report the case of a woman with breast cancer at an early age. Such malignancy is associated and was somehow induced by the rich family history, represented by the high prevalence of cancer in the ancestry. We report a 34-year-old woman with an extensive history of carcinoma in the family, who was diagnosed with breast cancer in July 2016. In order to confirm the diagnosis, it was required an ultrasound, which resulted in a 2.2×1.5 cm node on the right breast's left superior quadrant, classified as BIRADS 4A. It also performed an ultrasound-guided biopsy that showed a tubular carcinoma on the right breast with the following characteristics: positive for estrogen and progesterone receptor, positive for KI 67 (5%), and negative for HER2, with staging of T1cN0M0. During anamnesis, the patient mentioned menarche at 12 years old, history of birth control pills use for 10 years, no pregnancy, and no breastfeeding. When it comes to family history, a great number of relatives were previously diagnosed with some type of cancer. Her paternal grandfather had rectum cancer at 42 years old and breast cancer at 62 years old. The paternal grandmother passed away because of a fast-progression leukemia at the age of 68. It is important to mention that her progenitors were first cousins. Furthermore, the patient's dad was diagnosed with breast cancer at 62 years, alongside his three brothers who were also diagnosed with cancer: one with prostatic cancer at the age of 64 years and the other two with intestinal cancer at the ages of 64 and 68 years old. Considering such a family history, a genetic panel was performed, analyzing the genes related to hereditary cancer risk, and it identified mutations in the patient's *BRCA2* gene. Then, firstly, she performed a bilateral mastectomy in January 2017 with sentinel lymph node investigation, which was negative for neoplastic cells in the lymph nodes. Later, considering the BRCA2 mutation, in August 2017, the patient had to undergo prophylactic surgery: oophorectomy with salpingectomy.