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Blood Thicker Than Water: A Case Report on Familial Ovarian Cancer

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Abstract : Ovarian cancer is extremely hard to diagnose in its early stages, and those afflicted at the time of diagnosis are typically asymptomatic and in the late stages of the disease, with metastasis to other organs. Ovarian cancers often occur sporadically, with only 5% associated with hereditary mutations. Mutations in the BRCA1 and BRCA2 tumor suppressor genes have been found to be responsible for the majority of hereditary ovarian cancers. One type of ovarian tumor is Malignant Mixed Mullerian Tumor (MMMT), which is a very rare and aggressive type, accounting for only 1% of all ovarian cancers. Reported is a case of a 43-year-old G3P3 (3003), who came into our institution due to a 2-month history of difficulty of breathing. Family history reveals that her eldest and younger sisters both died of ovarian malignancy, with her younger sister having a histopathology report of endometrioid ovarian carcinoma, left ovary stage IIIb. She still has 2 asymptomatic sisters. Physical examination pointed to pleural effusion of right lung, and presence of bilateral ovarian new growth, which had a Sassone score of 13. Admitting Diagnosis was G3P3 (3003), Ovarian New Growth, bilateral, Malignant; Pleural effusion secondary to malignancy. BRCA was requested to establish a hereditary mutation; however, the patient had no funds. Once the patient was stabilized, TAHBSO with surgical staging was performed. Intraoperatively, the pelvic cavity was occupied by firm, irregularly shaped ovaries, with a colorectal metastasis. Microscopic sections from both ovaries and the colorectal metastasis had pleomorphic tumor cells lined by cuboidal to columnar epithelium exhibiting glandular complexity, displaying nuclear atypia and increased nuclear-cytoplasmic ratio, which are infiltrating the stroma, consistent with the features of Malignant Mixed Mullerian Tumor, since MMMT is composed histologically of malignant epithelial and sarcomatous elements. In conclusion, discussed is the clinic-pathological feature of a patient with primary ovarian Malignant Mixed Mullerian Tumor, a rare malignancy comprising only 1% of all ovarian neoplasms. Also, by understanding the hereditary ovarian cancer syndromes and its relation to this patient, it cannot be overemphasized that a comprehensive family history is really fundamental for early diagnosis. The familial association of the disease, given that the patient has two sisters who were diagnosed with an advanced stage of ovarian cancer and succumbed to the disease at a much earlier age than what is reported in the general population, points to a possible hereditary syndrome which occurs in only 5% of ovarian neoplasms. In a low-resource setting, being in a third world country, the following will be recommended for monitoring and/or screening women who are at high risk for developing ovarian cancer, such as the remaining sisters of the patient: 1) Physical examination focusing on the breast, abdomen, and rectal area every 6 months. 2) Transvaginal sonography every 6 months. 3) Mammography annually. 4) CA125 for postmenopausal women. 5) Genetic testing for BRCA1 and BRCA2 will be reserved for those who are financially capable.

Keywords : BRCA, hereditary breast-ovarian cancer syndrome, malignant mixed mullerian tumor, ovarian cancer
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