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Ovarian Localization of a Peripheral Primitive Neuroectodermal Tumor: Definitive Diagnosis is a Real Challenge

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Abstract **Case Report**

Primary neuroectodermal tumors (PNET) are aggressive tumors composed of small, round hyperchromic cells. Depending on their locations and the mother cell, they are classified into two subtypes: peripheral and central. Their locations in the genital tract are rare. We report here a case of peripheral primitive neuroectodermal tumor (pPNET) in the ovary, in a 42-year-old patient, whose definitive diagnosis was made on the basis of histological, immunohistochemical and molecular results.

Keywords: Neuroectodermal tumor, pPNET, ovary.

INTRODUCTION

Primary neuroectodermal tumor (PNET) is an aggressive tumor composed of small, round hyperchromic cells. It is grouped into 2 broad categories, those mimicking CNS neoplasms, namely central PNET, and those composed of small round cells with or without rosettes called extraosseous Ewing sarcoma or peripheral PNET (1). Its location in the genital tract is rare. We report here a case of peripheral primitive neuroectodermal tumor of the ovary whose definitive diagnosis was made on the basis of morphological and immunohistochemical and molecular results.

CLINICAL CASE

This is a 42-year-old woman, with no particular pathological history, presenting with deep abdominal pain lasting more than 2 months. Ultrasound and abdominal-pelvic magnetic resonance imaging (MRI) revealed a solido-cystic right ovarian mass, measuring 6 cm long axis. The patient underwent surgical intervention and an anatomopathological study. We received an operating specimen from a right adnexectomy almost completely, occupied by a solido-cystic mass (Figure A). It was conditioned for 24 hours in 4% buffered formol, then systematic samples were taken according to the standardized protocol.

An initial morphological study showed a malignant tumor proliferation with round cells within a desmoplastic stroma (Figure B). This led to a broad discussion on possible diagnoses. Immunohistochemistry subsequently showed intense and diffuse membrane expression of the anti CD99 antibody, moderate and diffuse cytoplasmic expression of the anti vimentin antibody, moderate and diffuse cytoplasmic expression of the anti NSE antibody, nuclear expression moderate in the 40% of tumor cells of the anti KI67 antibody and moderate and diffuse nuclear expression of the anti ERG antibody (Figure B). The tumor cells showed no expression for the following antibodies: anti chromogranin, anti synaptophysin, anti Desmin, anti WT1, anti myogenin, anti Melan1, anti TTF1, anti- Inhibin, anti calretinin, anti CD 30, anti FLI1, anti CK and anti OCT 4. The diagnosis of peripheral ovarian PNET was made and supported by a fluorescence in situ hybridization (FISH) study, looking for a translocation of the EWSR1 gene. The latter showed that 70% of cells are translocated.

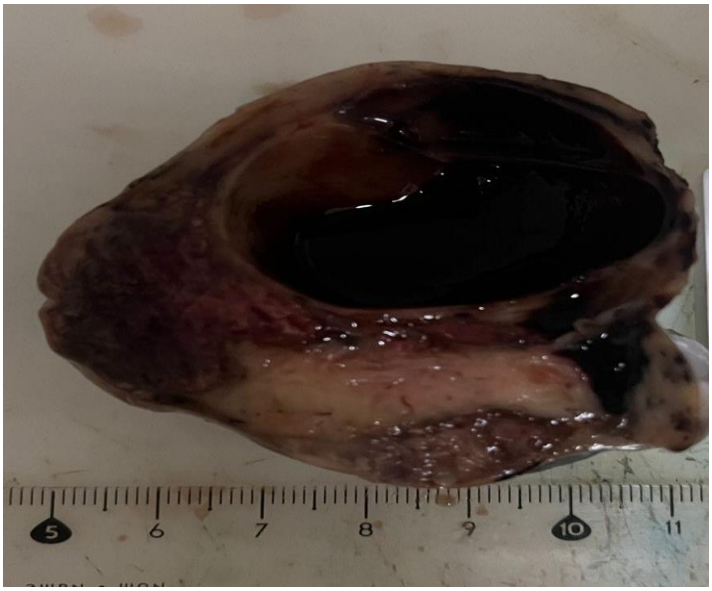

Figure A: Solido-cystic right ovarian mass, measuring 6 cm long axis

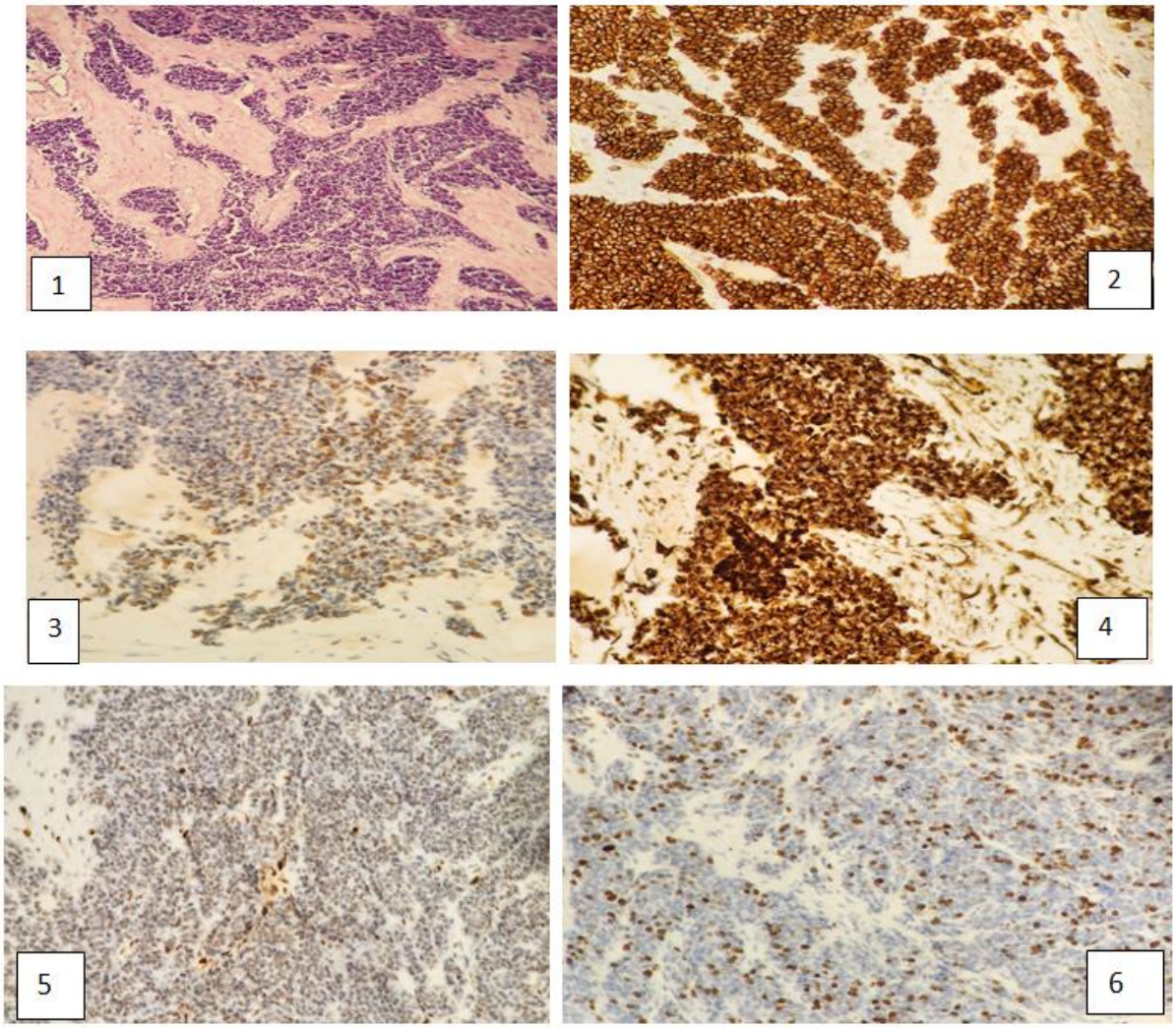

Figure B: 1) tumor proliferation with round cells within a desmoplastic stroma; 2) intense and diffuse membrane expression of the anti CD99 antibody, 3), moderate and diffuse cytoplasmic expression of the anti NSE antibody. 4) moderate and diffuse cytoplasmic expression of the anti vimentin antibody; 5) moderate and diffuse nuclear expression of the anti ERG antibody, 6) nuclear expression moderate in the 40% of tumor cells of the anti KI67 antibody

DISCUSSION

PNETs are the most common type of all ovarian neuroectodermal tumors [1]. They can be either peripheral type or central type. Central PNETs in the ovary originate from the nervous tissue present in ovarian teratomas [2] and are characterized by the negativity of the anti-CD99 antibody [3]. Peripheral PNETs at the ovarian level are less frequent than the central type [4], they are characterized by the presence of a proliferation of small round blue cells with diffuse and intense immunoreactivity for CD99 [2]. FLI1 and ERG are often expressed in the cases with the corresponding gene fusions [5]. Keratin expression is present in approximately in 25% of cases [5]. Other IHC markers that may also be expressed in pPNET include neuron-specific enolase (NSE), S100 protein, and vimentin. Genetically, all cases of Ewing sarcoma harbor a FET-ETS fusion. The most common Ewing sarcoma translocation, found in almost 85% of cases, is t(11;22)(q24;q12), resulting in EWSR1-FLI1 fusion transcript and protein. The second found in 10% of cases is t(21;22)(q22;q12), which gives EWSR1-ERG. The remaining cases have variant translocations that join either EWSFI1 or FUS (which, along with TAF15, form the FET family) to other members of the ETS family [5].

The tumor is frequently confused with other types of ovarian tumors such as granulosa cell tumor, hypercalcemic small cell carcinoma, and other tumors of hematopoietic and stromal origin. In our patient, the strong immunoreactivity for CD99 and the negativity of stromal, hematolymphoid epithelial and sex cord markers as well as the presence of a translocation of the EWSR1 gene in more than 70% of the tumor cells allowed us to rule out the above-mentioned tumors and to retain the diagnosis of an ovarian pPNET.

CONCLUSION

Primary neuroectodermal tumors in the gynecological tract are rare and pose a differential diagnosis problem, particularly in the ovary. Thus, faced with any hyperchromic round cell proliferation in the ovary, it is necessary to know how to evoke a PNET and confirm it as being of central or peripheral type by a combination of morphological evaluation, immunohistochemical analysis as well as a possible study molecular if possible.

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