Ginekol Pol. 2012, 83, 552-554

Rare case of uterine PEC-oma (Perivascular Epithelioid Cell Tumor) recurrence. Case report and literature review

Rzadki przypadek wznowy PEComa *(Perivascular Epithelioid Cell Tumor).* Przypadek kliniczny i przegląd piśmiennictwa

Issat Tadeusz[1,2], Maciejewski Tomasz[1], Beta Jarosław[1,2], Jakimiuk J. Artur[1,3]

[1] Department of Obstetrics and Gynecology, Central Clinical Hospital of Ministry of Interior and Administration, Warsaw, Poland,
[2] Department of Reproductive Health Research Institute of Mother and Child, Warsaw, Poland,
[3] Mossakowski Medical Research Centre Polish Academy of Sciences

Abstract

Perivascular epithelioid cell tumor (PEC-oma) is a rare mesenchymal neoplasm. Literature reports more than 100 cases of PEC-oma, a third of which is of uterine or uterine retroperitoneum origin. The case of a 59-year-old woman presented here is, to the best of our knowledge, the first described fast uterine PEC-oma recurrence of the tumor of the gastrointestinal tract origin. In this text the authors also present literature review concerning this rare female tumor.

Key words: **perivascular epithelioid cell tumor** / **PEC-oma** / **HMB 45** / **recurrence** /

Streszczenie

Perivascular epithelioid cell tumor (PEComa), są bardzo rzadkimi guzami pochodzenia mezenchymalnego. Do dnia dzisiejszego odnotowano w piśmiennictwie ponad 100 przypadków PEComa, z czego mniej więcej 1/3 występowała w macicy lub pokrywającej ją otrzewnej (retroperitoneum). Prezentowany przypadek 59 letniej pacjentki jest według nas pierwszym przedstawiającym szybką wznowę zmiany o typie PEComa zlokalizowaną w obrębie macicy o pierwotnym punkcie wyjścia z przewodu pokarmowego. W poniższym tekście przedstawiamy także przegląd piśmiennictwa dotyczący tego rzadkiego guza kobiecego narządu rodnego.

Słowa kluczowe: **perivascular epithelioid cell tumor** / **PEC-oma** / **HMB 45** / **wznowa** /

Corresponding author:
Artur Jakimiuk
Department of Obstetrics and Gynecology
Central Clinical Hospital of Ministry of Interior and Administration,
Woloska 137, 02-507 Warsaw, Poland,
tel. (0048) 225081120, fax. (0048) 225081125,
e-mail: jakimiuk@yahoo.coml

Otrzymano: **27.11.2011**
Zaakceptowano do druku: **11.06.2012**

Introduction

Perivascular epithelioid cell tumor (PEC-oma) is a rare mesenchymal neoplasm. In fact, histologically, the term PEC-oma describes a group of tumors and includes angiomyolipoma (AML), clear cell „sugar" tumors of the lung (CCST), lymphangioleiomyomatosis (LAM), clear cell myomelanocytic tumor of the falciform/ligamentum teres (CCMT) and adenocarcinomas of the pancreas, rectum, peritoneum, uterus, vagina and the heart [1]. Literature reports more than 100 cases of PEC-oma, a third of which are of uterine or uterine retroperitoneum origin [2]. The presented case, to the best of our knowledge, is the first described fast uterine PEC-oma recurrence of the tumor of the gastrointestinal tract origin.

Case Report

A 59-year-old woman was admitted to our hospital due to pelvic mass diagnosed at the ultrasound examination. History revealed hospitalization due to multiple peritoneal tumors 4 months prior to admission. Biphasic computer tomography examination showed polycyclic mass measuring 110x62mm with the intestine as a probable point of origin. Behind the uterus there was also a cyst, measuring 54x45mm. No ascites nor enlarged lymph nodes were observed. Other abdominal cavity organs i.e. liver, kidneys, adrenals, pancreas or spleen were normal. Subsequent pan-colonoscopy did not reveal any abnormalities. Serum biochemical tumor markers were negative (SCC-squamous cell carcinoma antigen, CA 19-9 (carbohydrate antigen 19-9), CYFRA 21.1, CEA (carcinoembryonic antigen), CA-125 (Cancer Antigen-125), CA-15,3 (Cancer Antigen-15,3). Serum levels of alpha-fetoprotein (AFP) were slightly above the normal range (10.1 IU/ml).

The patient was counseled and scheduled for surgical treatment. Laparotomy due to splenic flexure colon tumor suspicion was performed. Multiple abdominal masses were found – at the colon mesenterium, splenic flexure and greater omentum site. Additionally, one mass of pelvic location was noted. None of the tumors were infiltrating the surrounding tissues or organs. All tumors were surgically removed. Histopathological verification showed epithelioid atypical neoplastic cells with medium mitotic activity.

Additional immunohistochemical staining revealed HMB45 (Human Melanoma B-45) (+), Melan A (Melanocyte Antigen) (-), (Microphthalmia-associated Transcription Factor) MITF (+), SMA (Smooth Muscle Actin) (+), CALDESMON (+), DESMINA (+). The diagnosis of PEC-oma tumor was established on the basis of the clinical and histological findings. Postoperative period was uneventful and the patient was discharged in good overall condition. On day 59 after surgery she reported at the emergency department due to difficulty passing urine. Ultrasound examination performed at that time did not show any abnormalities and the patient was discharged home. Eighteen days after that episode (77 days since the surgery) she had computer tomography performed privately. Tomography revealed partially solid, partially cystic pelvic mass measuring 92x76mm. Solid component of the tumor increased the signal density after the contrast. The tumor displaced the bladder and the rectum with no signs of infiltration. There was no ascites, lymph nodes were not enlarged and abdominal organs appeared normal. Gynecological examination showed displacement of the cervix and palpable solid mass occupying the pelvis. Ultrasound examination revealed hyperechogenic mass within the uterus measuring as stated during tomography. Options of surgical treatment were discussed and total abdominal hysterectomy with bilateral salpingo-oophorectomy was scheduled. Second laparotomy showed massive solid adhesions of the uterus, adnexa and bowel loops. Uterus was of normal size with the tumor originating from the posterior wall being located partially within recto-vaginal septum. There were no macroscopic abnormalities within ovaries. The procedure was uneventful and the histological studies of the provided specimen revealed mesenchymal tumor with necrotic foci. Additional immunohistological staining showed positive reaction for SMA (+), DESMINA and HMB45 and MIB1 (mitiotic index, prolferative antigen Ki67) in around 10% of specimen cells. PEC-oma was again confirmed in the final diagnosis.

Discussion

The name perivascular epithelioid cells (PEC) was established in 1992 by Bonetti et al., who described morphemic and immunohistochemical characteristics of the tumor cells [3]. In 1996 Zamboni et al., proposed the name for the whole group of tumors including already mentioned CCST, CAM, LAM and AML. The WHO definition published in 2002 states that PEComa are mesenchymal tumors containing perivascular epithelioid cells of defined morphology and immunohistochemistry. Phenotypically, those tumors express on their surface melanogenesis particles i.e. HMB-45, Melan-A (MART-1, melanoma associated antigen), centrally staining actin, desmin, vimentin and do not express particles of epithelium like cytokeratin. Unlike cells of clarocellular sarcoma, PEC-oma does not express S100 particles. Another characteristic is abnormal vascularization of the tumor (LAM, AML) [5]. Taking into account higher prevalence of PEC-oma in women there was a hypothesis linking the tumor with hormonal imbalance. Some studies showed the presence of the progesterone and more dominant estrogen receptors on the surface of the PEC-oma tumors [6,7].

Further studies showed that CD-1a particle present in the PEC-oma cells helps in differentiating the mass from the epithelioid smooth muscle tumors [8].

In the literature there are studies showing coincidence of the PEC-oma and tuberous sclerosis complex (TSC), autosomal dominant disease being characterized by epilepsy, mental retardation and multiple tumors [6,9]. More recent studies did not support this hypothesis.

The diagnosis of PEC-oma is relatively rare. Second most common after the uterus tumor site is gastrointestinal tract. As far as the last localization is concerned, the differential diagnosis involves melanoma, GIST (Gastrointestinal Stromal Tumors), clarocellular tumors of soft tissues, sarcomas, leyomyosarcomas and ganglioma. The malignancy potential, growing dynamics or patterns of PEC-oma tumors has not been clearly defined. Literature data, however, supports the hypothesis that it is more prevalent in young women [10]. This kind of tumor can be located in different organs with the uterus being the most frequent one [6, 11]. One study showed that age at the time of diagnosis ranges from 17 to 79 years, with the average age at 54 [12]. The most common symptoms were uterine bleeding, cramps or growing mass in the hypogastric area.

The prediction of the dynamics of the PEC-oma tumors appears difficult. Some patients face metastases, recurrence or fatal course of the disease even within one or two years since the surgical treatment [6,13]. Some studies suggested sub-dividing the group into three subgroups depending on the prognosis [1]. The so-called benign group includes masses measuring less than 5 cm with low number of mitotic divisions (1<50HPF), absence of multinuclear cells or cells with excessive cytoplasm. Two other sub-groups are tumors with uncertain and high malignancy potential, respectively, being characterized by multinuclear pleomorphic cells.

In most cases surgery seems to be the treatment of choice. However, some patients may benefit from the complementary therapy. Some studies described cases of metastatic PEC-oma with the course of doxorubicin and ifosphamide followed by radiotherapy [6] or only chemotherapy with ifosphamide following surgical procedure [10]. One study presented the case of a 9-year-old patient with uterine PEC-oma with lymph nodes metastases being treated by surgery followed by chemotherapy (ifosphamide, vincristine, and doxorubicin) and radiotherapy [1]. Authors of the study claimed remission of the disease with the acute lymphocytic leukemia diagnosed 4 years after aggressive treatment for PEC-oma [14]. One study presented the case of PEC-oma recurrence in lungs 7 years after primary surgical removal of the uterine mass [15].

In conclusion, present data is insufficient to provide reliable algorithms for PEC-oma treatment strategies. The presented case and literature data support the theory that even though surgical treatment appears to be primary and only choice, it would be reasonable to consider complementary therapy and very close follow-up.

References

1. Folpe A, Mentzel T, Lehr H, [et al.]. Perivascular epithelioid cell neoplasms of soft tissue and gynaecologic origin: a clinicopathologic study of 26 cases and review of the literature. *Am J Surg Pathol.* 2005, 29, 1558-1575.
2. Fadare O. Perivascular epithelioid cell tumor (PEComa) of the uterus: an outcome-based clinicopathologic analysis of 41 reported cases. *Adv Anat Pathol.* 2008, 15, 63-75.
3. Bonetti F, Pea M, Martignoni G, Zamboni G. PEC and sugar. *Am J Surg Pathol.* 1992, 16, 307-308.
4. Zamboni G, Pea M, Martignoni G, [et al.]. Clear cell "sugar" tumor of the pancreas. A novel member of the family of lesions characterized by the presence of perivascular epithelioid cells. *Am J Surg Pathol.* 1996, 20, 722-730.
5. Sharma S, Kotru M, Gupta R. PEComata: highly melanotic multiple perivascular epithelioid cell tumors (PEComa) of the uterus. *APMIS.* 2008, 116, 1000-1003.
6. Bonetti F, Martignoni G, Colato C, [et al.]. Abdominopelvic sarcoma of perivascular epithelioid cells. Report of four cases in young women, one with tuberous sclerosis. *Mod Pathol.* 2001, 14, 563-568.
7. Martignoni G, Pea M, Reghellin D, [et al.]. Perivascular epithelioid cell tumor (PEComa) in the genitourinary tract. *Adv Anat Pathol.* 2007, 14, 36-41.
8. Fadare O, Liang S. Epithelioid smooth muscle tumors of the uterus do not express CD1a: a potential immunohistochemical adjunct in their distinction from uterine perivascular epithelioid cell tumors. *Ann Diagn Pathol.* 2008, 12, 401-405.
9. Folpe A, Goodman Z, Ishak K, [et al.]. Clear cell myomelanocytic tumor of the falciform ligament/ligamentum teres: a novel member of the perivascular epithelioid clear cell family of tumors with a predilection for children and young adults. *Am J Surg Pathol.* 2000, 24, 1239-1246.
10. Jeon I, Lee S. Multimodal treatment using surgery, radiotherapy, and chemotherapy in a patient with a perivascular epithelioid cell tumor of the uterus. *J Pediatr Hematol Oncol.* 2005, 27, 681-684.
11. Fukunaga M. Perivascular epithelioid cell tumor of the uterus: a case report. *Int J Gynecol Pathol.* 2004, 23, 287-291.
12. Fink D, Marsden D, Edwards L, [et al.]. Malignant perivascular epithelioid cell tumor (PEComa) arising in the broad ligament. *Int J Gynecol Cancer.* 2004, 14, 1036-1039.
13. Greene L, Mount S, Schned A, Cooper K. Recurrent perivascular epithelioid cell tumor of the uterus (PEComa): an immunohistochemical study and review of the literature. *Gynecol Oncol.* 2003, 90, 677-681
14. Jeon I, Yi D. Acute lymphoblastic leukemia secondary to chemoradiotherapy for perivascular epithelioid cell tumor of uterus. *Pediatr Hematol Oncol.* 2009, 26, 85-88.
15. Dimmler A, Seitz G, Hohenberger W, [et al.]. Late pulmonary metastasis in uterine PEComa. *J Clin Pathol.* 2003, 56, 627-628.