MedNEXT
Journal of Medical an Health Sciences

CASE REPORT

DOI: 10.54448/mdnt23201

Perivascular malignant epithelioid cell tumor (PEComa) of the uterus: a case report

Cibele Alexandra Ferro[1], Maria Fernanda Roncolato Catarino da Fonseca Pereira[1], Maria Júlia de Andrade Parizati[1], Ana Clara Figueiredo de Faria[1], Anaisa Renata Michelino[1], Bárbara Nascimento Mendes[1], Brunna Maria Andrade Ribeiro[1], Caroline Aparecida Martins Bezerra[1], Julia Espolador Bianchi[1], Laíne Ribeiro Antonelli[1], Manuela Lopes Poiati[1], Guilherme Spagna Accorsi[2]*

[1] FAMECA - Faculty of Medicine of Catanduva/Centro Universitário Padre Albino (UNIFIPA), Catanduva, São Paulo, Brazil.
[2] FAMECA - Faculty of Medicine of Catanduva/Centro Universitário Padre Albino. Gynecology and Obstetrics department, Catanduva, São Paulo, Brazil.

*Corresponding author: Dr. Guilherme Spagna Accorsi, FAMECA - Faculty of Medicine of Catanduva/Centro Universitário Padre Albino (UNIFIPA), Catanduva, São Paulo, Brazil. Email: accorsi_gui@hotmail.com

DOI: https://doi.org/10.54448/mdnt23201

Received: 12-12-2022; Revised: 02-27-2023; Accepted: 03-07-2023; Published: 03-08-2023; MedNEXT-id: e23201

Abstract

Perivascular epithelioid cell tumors, called PEComas, have a mesenchymal origin with immunoreactivity for melanocytic and smooth muscle markers. Its incidence in the form of uterine involvement is rare, between 1 and 2 cases per million inhabitants, affecting mainly women in their fifth decade of life. The present study was elaborated according to the rules of the CARE case report. The patient's medical record was analyzed, and who authorized access to it and signed the Free and Informed Consent Form, together with those involved in this work. This patient underwent treatment and medical follow-up with the supervisor of this work. Therefore, this study aimed to describe a rare clinical case report of uterine malignant perivascular epithelioid cell tumor (PEComa). Although the literature on this subject is scarce and there are no consistent criteria for diagnosis and treatment, our case in question presented aspects of unfavorable evolution (a large number of cytological atypia and high mitotic index) characterizing a PEComa with uncertain malignant potential, requiring a treatment adjuvant after surgery. The patient evolved well after the surgery and adjuvant treatment, undergoing quarterly follow-ups at the oncology and gynecology outpatient clinic, with a physical examination and nuclear magnetic resonance of the pelvis for control purposes.

Keywords: Malignant tumor. Perivascular epithelioid cells. PEComa. Uterus.

Introduction

Perivascular epithelioid cell tumors, called PEComa, have a mesenchymal origin with immunoreactivity for melanocytic and smooth muscle markers [1,2]. Its incidence in the form of uterine involvement is rare, between 1 and 2 cases per million inhabitants, affecting mainly women in their fifth decade of life [1,3,4]. The etiology and risk factors are still uncertain, although studies relate the origin of PEComas to the appearance of neural stem cells in the lining of vessels that could differentiate into myocytes or melanocytic cells [5].

These tumors are often found in the retroperitoneum, uterus, cervix, or gastrointestinal tract, and in some cases, soft tissues and bones [1,4,6]. The most common symptoms include vaginal bleeding, abdominal pain, and uterine enlargement. The gold standard diagnosis is a biopsy together with immunohistochemical analysis [7]. Surgical treatment is the most indicated and may be associated with adjuvant treatment with radiotherapy.

Therefore, this study aimed to describe a rare clinical case report of uterine perivascular malignant epithelioid cell tumor (PEComa).

Methods

Study Design

The present study was elaborated according to the rules of the CARE case report (https://www.care-statement.org/). A descriptive literature review was also

carried out to provide sufficient scientific data for the theoretical basis of this study. The patient's medical record was analyzed, and who authorized access to it and signed the Free and Informed Consent Form, together with those involved in this work. The patient underwent treatment and medical follow-up with the supervisor of this work. A thorough evaluation of the aspects inherent to the physical examination, complementary exams, and surgical technique was carried out, to correlate with the literature cited in the bibliography. Data collection and analysis of the patient's medical records were carried out at Hospital Escola Emílio Carlos, Catanduva, São Paulo, Brazil.

Ethical Approval

This study was analyzed and approved with the number 5.930.073 by the Research Ethics Committee from the FAMECA / UNIFIPA, Catanduva, Brazil, and obtaining the Informed Consent Form according to CNS/CONEP Resolution 466/12.

Case Report

Patient Information and Clinical Findings, Timeline, Diagnostic Assessment, Therapeutic Intervention, and Follow-up

A woman, 59 years old, hypertensive and dyslipidemic, attends an oncology and gynecology outpatient clinic complaining of daily bleeding with the oscillating flow for 1 year and a half - without the need for blood transfusion, after a 5-year break (menopause at age 54). She denies dysmenorrhea, dysuria, dyspareunia, and gastrointestinal tract disorders. Upon speculum examination, the cervix can be visualized, as the presence of active bleeding in a small amount from the external orifice, the presence of bleeding in the cul-de-sac, and the absence of lesions or ectopy. On vaginal examination, the cervix was atrophic, the uterus mobile, the cervix mobilization painless, and non-palpable appendages.

Because of the alterations found in the physical examination and the symptomatic complaint of the patient, a uterine curettage and magnetic resonance imaging of the pelvis was requested. The anatomopathological product of the uterine curettage revealed a poorly differentiated malignant neoplasm, invasive amid blood clots.

Also, magnetic resonance imaging of the pelvis showed a heterogeneous signal in the myometrium due to nodules suggestive of intramural leiomyomas with a submucosal component located in the posterior fundal region measuring 2 cm. Inside the uterine cavity, there is a heterogeneous expansive formation, measuring about 4.2x2.7 cm, extending from the body to the internal orifice of the uterine cervix (which may represent a primary endometrial neoplasm).

Figure 1. Sagittal section of magnetic resonance imaging of the pelvis: heterogeneous expansive formation, measuring about 4.2 x 2.7 cm, extending from the body to close to the internal orifice of the uterine cervix.

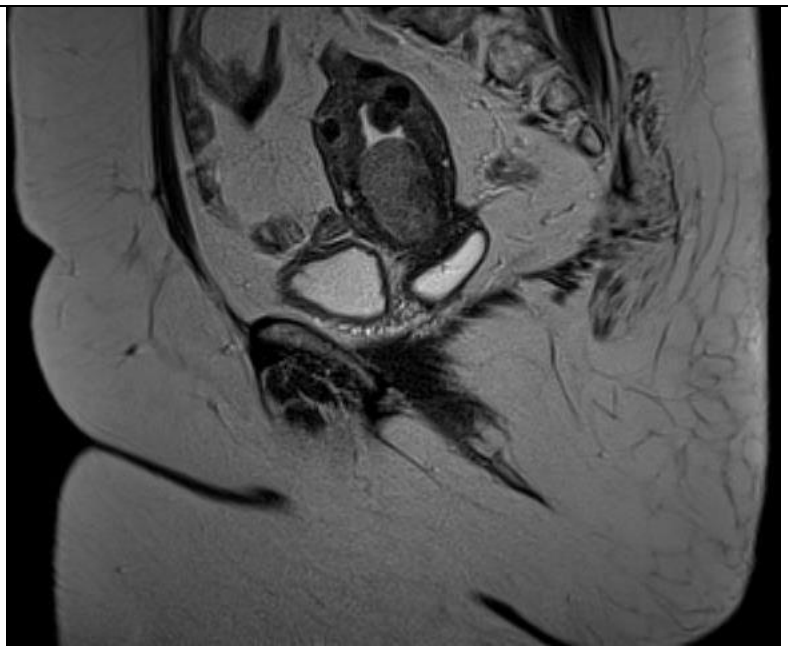
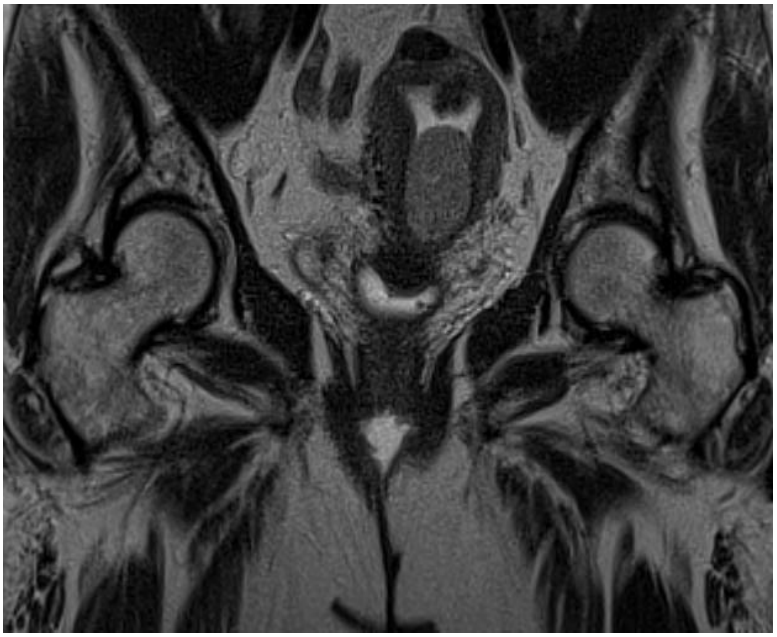


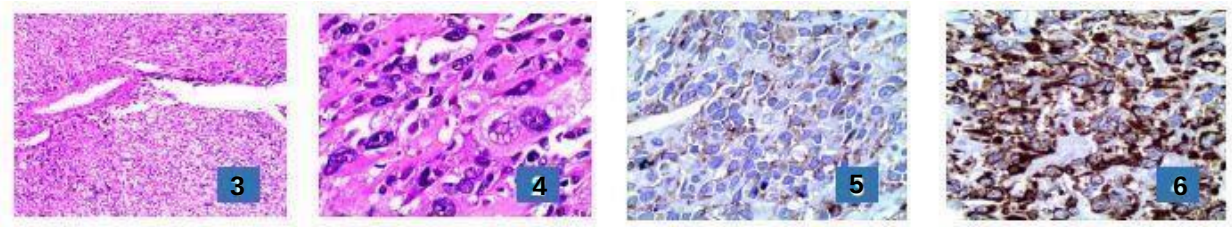
Figure 2. Coronal section of magnetic resonance imaging of the pelvis: heterogeneous expansive formation, measuring about 4.2 x 2.7 cm, extending from the body to close to the internal orifice of the uterine cervix.



Based on these findings, an elective hospitalization was performed to perform a laparoscopic hysterectomy + bilateral salpingo-oophorectomy + sentinel lymph node. In the inventory of the cavity, it is possible to visualize a uterus with little increase in volume, annexes without alterations and a cavity without implants. After dissecting the pelvic spaces, sentinel lymph nodes were not identified bilaterally. Pelvic lymphadenectomy was performed respecting the anatomical limits. The patient was discharged on the first postoperative day without complaints.

The product of total hysterectomy and bilateral salpingo-oophorectomy confirmed poorly differentiated mesenchymal neoplasm, with fusiform areas and epithelioid areas, submucosal and pedunculated, measuring 4.0 x 3.0 cm in the largest axes and protruding into the endocervical canal. Blood, lymphatic or perineural vascular neoplastic invasion was not detected. Submucous leiomyoma measuring 1.5 cm in the longest axis and located in the uterine fundus region. Absence of metastases in the 13 examined lymph nodes. Immunohistochemistry complemented the diagnosis with positive AML, positive CD10, positive desmin, positive weak and focal melan-A, positive estrogen receptor and positive progesterone receptor – complementing the diagnosis of uterine PEComa with compatible findings (**Figures 3-6**).

Figure 3. High epithelioid and Fusoepithelioid cellularity of microvacuolated clear cells with scarce stroma and the presence of small and medium-sized vascular ramifications. **Figure 4.** Significant nuclear atypia of anisokaryosis and hyperchromasia type. **Figure 5.** High mitotic activity with up to 09 mitoses per 10 CGA and with atypical mitoses. **Figure 6.** Undetected necrosis.



After surgery, the patient was referred for radiotherapy, which proposed the performance of 3D conformational radiotherapy in pelvic drainage fields with 45 Gy in 25x1.8 Gy/day in 25 sessions and at the

end of this treatment, the patient underwent 4 brachytherapy sessions in São José do Rio Preto. The patient evolved well, without intercurrences and other complaints.

Discussion

PEComas can be classified into benign and malignant. For a PEComa to be considered malignant, some criteria are defined: size > 5 cm, infiltrative growth, high nuclear grade and cellularity, mitoses > 1/50 HPFs, necrosis, and lymphovascular invasion [1]. The classification by the World Health Organization carried out in 2002 denominated the representative diseases of PEComa in lymphangioleiomyomatosis, angiomyolipoma, and lung clear cell tumor [8,9]. Furthermore, there are two distinct patterns identified: epithelioid – representing 100% of cases, and fusiform – present in 37% of cases [2].

Also, PEComas are associated with mutations in the TSC1 and TSC2 genes, in addition to the rearrangement of the TFE3 gene, which has a predominant epithelioid morphology with clear cells and positive staining for melanocytic markers such as HMB-45, with aggressive behavior in 52% of cases [2,10]. Thus, despite being rare and having common symptoms with differential diagnoses of other tumors, they may have an aggressive prognosis when diagnosed late.

Conclusion

Although the literature on this subject is scarce and there are no consistent criteria for diagnosis and treatment, our case in question presented aspects of unfavorable evolution (a large number of cytological atypia and high mitotic index) characterizing a PEComa with uncertain malignant potential, requiring a treatment adjuvant after surgery. The patient evolved well after the surgery and adjuvant treatment, undergoing quarterly follow-ups at the oncology and gynecology outpatient clinic, with a physical examination and nuclear magnetic resonance of the pelvis for control purposes.

Acknowledgement

Not applicable.

Funding

Not applicable.

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Informed consent

Was applied.

Data sharing statement

No additional data are available.

Conflict of interest

The authors declare no conflict of interest.

Similarity check

It was applied by Ithenticate@.

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