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A Rare Case Report of Perivascular Epithelioid Cell Neoplasm (PEComa) In A 54-Year-Old Woman With Endometrial Polyp

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Abstract

Endometrial polyps are a common cause of abnormal uterine bleeding in postmenopausal patients. The endometrial polyps, an abnormal growth containing glands, stroma, and blood vessels, are usually benign. However, an increased risk of malignancy occurs in postmenopausal bleeding, hereditary cancer syndrome, and tamoxifen use. Perivascular epithelioid cell tumors (PEComas) are rare mesenchymal tumors characterized by perivascular epithelioid cells exhibiting features of both melanocytes and smooth muscle cells. These tumors can manifest as benign, with uncertain or malignant potential. Only 114 cases of gynecologic PEComas have been reported, primarily affecting the uterus, cervix, vagina, adnexa, broad ligament, and vulva. However, no previous reports of PEComa are arising from endometrial polyps. Here, we present the case of a postmenopausal woman undergoing hormone replacement therapy (HRT) who experienced unscheduled vaginal bleeding. Following office, no-touch hysteroscopic removal of an endometrial polyp, a PEComa tumor was incidentally discovered. After a thorough discussion with the patient, a decision was made to pursue a follow-up plan without additional surgery. Two years later, the patient remained in a satisfactory condition, and two subsequent endometrial biopsies showed no remarkable changes. While the occurrence of endometrial polyps and PEComa can be concurrent or independent, managing such cases presents potential challenges.

Keywords: Perivascular Epithelioid Cell Neoplasm (PEComa), endometrial polyps, postmenopausal vaginal bleeding, Office no-touch hysteroscope

Introduction

Perivascular epithelioid cell neoplasms (PEComa) are rare mesenchymal tumors. The existence of perivascular epithelioid cell neoplasms dates to 1991; the term "PEComa" was introduced by Zamboni et al. in 1996 **[1]**. In 2002, the first uterine perivascular epithelioid cell neoplasms (PEComas) were reported **[2]**. PEComas are groups of perivascular epithelioid cells with features of melanocytes and smooth muscle cells. PEComas are combined groups of tumors that include: 1) angiomyolipomas (AMLs), 2) clear-cell sugar tumors, primary extrapulmonary sugar tumors (PEST), 3) lymphangioleiomyomatosis (LAM), 4) clear-cell myomelanocytic tumor (CCMT) of the falciform ligament/ligamentum teres, 5) primary cutaneous PEComa (CCCMT-cutaneous clear cell myomelanocytic tumor) and 6) PEComas not otherwise specified (NOS). **[3]** PEComas-NOS have been reported in almost every body site. The uterus is the most said site. 114 women have been documented in the English literature, with one-third of these cases affecting the uterus. **[4]** The

diagnosis is based on the positive staining expression for smooth muscle antigen and the melanocyte antigen, HMB-45, in these cells. **[4]** Typically, these tumors are rare and predominantly benign. Folpe et al. divided the tumors into mild, uncertain malignant potential, or malignant categories. **[6]** Gynecological PEComa primarily occurs in the uterine body or cervix, vagina, and adnexa. The most common complaint is vaginal bleeding. No previous report of PEComa arises from endometrial polyps.

Due to the scarcity of reported clinical cases of PEComa, no standardized protocols for diagnosis and treatment have been established. Therefore, it is crucial to describe each issue individually. In this report, we present a case of a postmenopausal woman who was undergoing hormone replacement therapy (HRT) but experienced unscheduled vaginal bleeding. Following the hysteroscopic removal of an endometrial polyp, a PEComa tumor was discovered.

Case History:

A 54-year-old woman was seen at our gynecology clinic due to HRT for 1 year with unscheduled vaginal bleeding for two weeks.

One year before this episode, the patient was evaluated at the clinic for osteoporosis and menopausal symptoms. Her last period ended at 52. Divigel (estradiol gel providing 0.25 mg estradiol) and oral progestin (100 mg progesterone) per day were given at our clinic.

Six months before this presentation, unexpected vaginal bleeding occurred. Transvaginal sonography revealed the presence of a small uterine fibroid measuring 1.6x1.7 cm on the posterior wall, with an endometrial thickness of 7.37 mm. Following a diagnostic dilation and curettage (D&C) procedure was performed, the pathology report indicated a proliferative phase.

Afterward, the patient resumed hormone replacement therapy (HRT). During this episode of vaginal bleeding, the amount was sporadic and relatively small, accompanied by minor blood clots for two weeks. However, there were no signs of abdominal pain, distention, or palpable masses.

The patient's medical history revealed that she was healthy and single with no pregnancy history and no clinical or family history of tuberous sclerosis or skin cancer. Her yearly Pap tests consistently showed average results.

The patient's body mass index (BMI) was measured at 20 (156.7 cm/49.2 kg) upon physical examination. Her blood pressure was 121/69 mmHg, and her pulse rate was 82 beats per minute. Transvaginal sonography revealed a uterine size and endometrial thickness of 8 mm, with the small uterine fibroid remaining unchanged. No other abnormalities were observed during the examination.

To address the situation, an office no-touch hysteroscopic surgery was performed. A rigid hysteroscope (RUDOLF Medical Germany) with a diameter of 4.3 mm was used, along with a continuous-flow sheath and a separate 5 Fr operating sheath. Within the procedure, a small endometrial polyp was discovered, and it was subsequently removed and biopsied using a grasper and scissors under direct visualization. After removing endometrial polyps, a manual vacuum aspiration (MVA) endometrial biopsy was performed.

The histological examination of the polypectomy and endometrial tissue specimen consisted of multiple tissue fragments weighing a total of 0.49 grams.

The detailed microscopic analysis demonstrated that the H&E section exhibited fragmented soft tissue containing a mixture of dilated thin and thick-walled vascular channels. Within this tissue, there were intervascular spindled and epithelioid cells present. These cells displayed moderate pale to eosinophilic cytoplasm and vesicular nuclei. Notably, there was no evidence of nuclear pleomorphism or hyperchromasia. SMA (smooth muscle actin) and HMB-45 expression in these cells were heterogeneous. Mitotic figures were scarce, and no coagulative necrosis was observed. Based on these findings, a diagnosis of perivascular epithelioid cell tumor (PEComa) of the uterus is suggested. After examining the pathological findings, few mitotic figures were present, and no signs of coagulative necrosis. Following a thorough discussion with the patient, it was decided to proceed with a follow-up plan without surgery. Subsequently, two years later, the patient's condition remained satisfactory, and there were no signs of recurrence as the two endometrial biopsies showed unremarkable change.

Discussion

Endometrial polyps are among the most common causes of abnormal genital tract bleeding in women. Most endometrial polyps are benign. However, an increased risk of malignancy occurs in selected patients with postmenopausal bleeding and tamoxifen use. Uterine endometrial polyps associated with PEComa have never been reported. PEComa arises from perivascular epithelioid cells and occurs in various organs such as the uterus, retroperitoneum, kidney, and gastrointestinal tract and exhibits potential for malignant behavior. Gynecological PEComas account for just over one-fourth of the overall PEComa cases reported in the literature. Surgery is the most recommended primary treatment, while adjuvant therapy is generally reserved for high-risk patients. **[5]**

The most sensitive marker to diagnose PEComa is HMB-45, and nearly all PEComas were found to be positive staining for HMB-45. In agreement with a previous systematic review published in 2019. **[4]** Except for HMB-45, other indicators included tyrosinase, microphthalmia transcription factors, NKI/C3, and smooth muscle markers, such as smooth muscle actin (SMA), desmin, h-caldesmon, pan-muscle actin, muscle myosin, and calponin, etc. **[7]**

The pathogenesis of PEComas is still unknown but, in some cases, be associated with the genetic condition tuberous sclerosis complex (TSC), of which mutations in the TSC1 or TSC2 genes. Mutations in the TSC1 or TSC2 genes lead to the overactivation of the mTOR pathway, which causes increased cell growth, blood vessel formation, and protein synthesis. An overactive mTOR pathway is also thought to be involved in some sporadic cases of PEComas. These patients may benefit from an mTOR inhibitor when these genetic mutations are present.

Signs and symptoms of PEComas vary between patients and tumor location. For women with gynecologic PEComas in the reproductive tract, these tumors may present with painful or painless mass with vaginal bleeding. Among the patients with PEComa of the gynecologic tract, the uterus was reported to be the most affected anatomic site (58.6 %), followed by the cervix (10.5 %), while only 1 case occurred in the vulva **[4]**. Some PEComas cause no symptoms and are found incidentally when patients undergo imaging for other reasons. In our case, the only sign was painless vaginal bleeding. PEComas-NOS have been reported in almost every body site. Except for the uterus, other standard areas include the genitourinary tract, gastrointestinal tract, and retroperitoneum. PEComas have also been reported in the oral cavity, the orbit, and the skull base. **[8]**

Endometrial polyps are benign growths in the uterine lining. Although they may be malignant transformations, they can cause symptoms such as abnormal uterine bleeding or infertility. While endometrial polyps and PEComa can both occur simultaneously or independently, there exhibits a potential for management challenges.

No reports confirm the relationship between female hormones and PEComas in women. While our case was positive staining for HMB-45 and SMA, mitotic figures were scarce, and no coagulative necrosis was observed.

We decided to follow up without other surgical intervention and continued to use HRT for this patient. The follow-up period in our case is limited (24 months) and just one case. Although the optimal treatment of gynecological PEComas is controversial, surgical resection remains the cornerstone. Adjuvant therapy is suggested in high-risk patients to increase disease control. A multidisciplinary approach should be critical in treatment decision-making regarding gynecological PEComas.

Clinically close follow-up is necessary for all patients diagnosed with gynecological PEComas, even patients with low-risk features such as our case.

In conclusion, a rare endometrial polyp with PEComa case was reported, and the diagnosis and management were discussed. However, further reports of similar cases are needed.

Contribution of Authorship

CSY initiated the idea of the subject. CSY and YLL performed a literature search on the subject. YLL YSB collects the data. All authors approved the final version of this article.

All authors followed The Declaration of Helsinki: ethical principles for medical research involving human subjects. (World Medical Association):

Disclosure of Interests: The authors of this article have no conflict of interest to disclose.

References

1. Pea M, Bonetti F, Zamboni G, Martignoni G, Riva M, et al. (1991) Melanocyte-marker-HMB-45 is regularly expressed in angiomyolipoma of the kidney. Pathology. 23(3): 185–188.
2. Vang R, Kempson RL (2002) Perivascular epithelioid cell tumor ('PEComa') of the uterus: a subset of HMB-45-positive epithelioid mesenchymal neoplasms with an uncertain relationship to pure smooth muscle tumors. Am J Surg Pathol. 26(1): 1–13.
3. Sobiborowicz A, Świtaj T, Teterycz P, Spałek MJ, Szumera-Ciećkiewicz A, et al. (2021) Feasibility and Long-Term Efficacy of PEComa Treatment-20 Years of Experience. J Clin Med. 10(10): 2200.
4. Liu CH, Chao WT, Lin SC, Lau HY, Wu HH, et al. (2019) Malignant perivascular epithelioid cell tumor in the female genital tract: Preferred reporting items for systematic reviews and meta-analyses. Medicine. 98(2): e14072.
5. Musella A, De Felice F, Kyriacou AK, Barletta F, Di Matteo FM, et al. (2015) Perivascular epithelioid cell neoplasm (PEComa) of the uterus: A systematic review. Int J Surg. 19: 1-5.
6. Folpe AL, Mentzel T, Lehr HA, Fisher C, Balzer BL, et al. (2005) Perivascular epithelioid cell neoplasms of soft tissue and gynecologic origin: a clinicopathologic study of 26 cases and review of the literature. Am J Surg Pathol. 29(12): 1558–1575.
7. Fadare O (2008) Uterine PEComa: appraisal of a controversial and increasingly reported mesenchymal neoplasm. ISSO. 5: 7.
8. Alam MS, Mukherjee B, Krishnakumar S, Biswas J (2017) Malignant perivascular epithelioid cell tumor of the orbit: Report of a case and review of literature. Indian J Ophthalmol. 65(9): 889-891.