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Case Report

47 Year Old Female with a Recurrent Abdominal Perivascular Epithelioid Cell Tumor: Case Report and Literature Review

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Abstract: Perivascular epithelioid cell tumors (PEComas) are rare mesenchymal neoplasms characterized by histologically and immunohistochemically distinctive epithelioid or spindle cells. These tumors express both smooth muscle markers and melanocytic markers. They can arise in various anatomical locations throughout the body, including the gastrointestinal tract, genitourinary system, retroperitoneum, and soft tissues. The exact cell of origin for these tumors remains unidentified. Clinical outcomes vary widely, spanning benign to malignant behavior, with the potential for local recurrence and metastasis. The management of PEComas typically involves surgical resection, though use of adjuvant chemotherapy and immunotherapy has been reported. The high recurrence rate of PEComas highlights the necessity for long-term surveillance and a multidisciplinary approach to treatment. We present the case of a 47-year-old female with a previous abdominal PEComa, who presented to our clinic with an enlarged recurrent abdominal tumor. Confirmation of recurrence was obtained through ultrasound and PET-CT imaging. Additionally, post-operative pathology findings supported the diagnosis.

Keywords: Perivascular epithelioid cell neoplasms, Recurrence, Mesenchymal neoplasms

1. INTRODUCTION

Perivascular epithelioid cell tumors (PEComas), are mesenchymal neoplasms made up of distinctive epithelioid or spindle-shaped cells. These tumors are identified by their unique histological and immunohistochemical features, with the cells testing positive for markers associated with both smooth muscle (e.g., actin and desmin) and melanocytic cells (e.g., HMB-45 and Melan-A).[1,2] These are rare tumours that can develop in various parts of the body, including but not limited to the gastrointestinal tract, genitourinary system, retroperitoneum, and soft tissues.[3] The specific cell type that gives rise to these tumors is unknown. Perivascular epithelioid cells are not typically found, and the name describes the tumor's appearance when examined under a microscope, it is thought to originate from a unique perivascular cell type that exhibits myomelanocytic differentiation.[4] The most common tumors within the PEComa group are renal angiomyolipoma and pulmonary lymphangioleiomyomatosis, both of which occur more frequently in individuals with tuberous sclerosis complex.[5] Additionally, many types of PEComas show a higher incidence in females.

The clinical behavior of these tumors is variable, ranging from benign to malignant, with potential for local recurrence and metastasis.[6] The management of PEComas often involves surgical resection, surgical resection with clear margins remains is the prefered manegment.[7] For lesions exhibiting malignant features not amenable to operative removal, use of adjuvant chemotherapy and immunotherapy has been reported but there is still limited data involving these therapies, though targeted therapies, such as mTOR inhibitors, have shown promise in some cases. Prognosis depends on several factors, including tumor size, location, and histological features indicative of aggressive behavior.[8]

2. CASE REPORT

A 47 year woman came to the outaptient clinic complaning from abdominal pain, fatigue, changes in bowel habits and weight gain for the last few

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months. The patient has a history of PEComa, an abdominal mass diagnosed at another hospital two years ago. At that time, Informed consent was obtained from the patient and surgery was performed to remove the mass, and the procedure also included the removal of the uterus and ovaries.

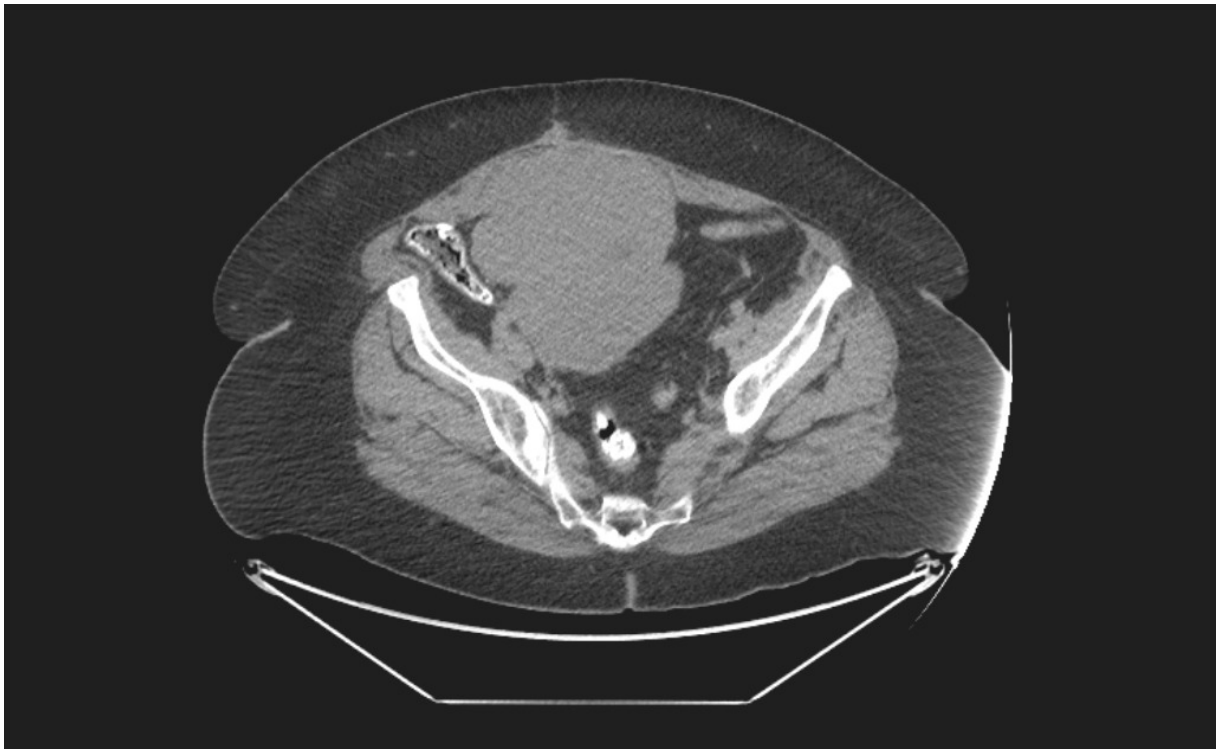
The patient had a recurrence of the tumor one month prior to her visit to our hospital. This recurrence was initially recognized by the urology department at a different hospital following an abdominal ultrasongraphy (USG) performed due to a urinary tract infection (UTI) the patient had at that time.

The USG report describes a heterogeneous hypoechoic solid mass with a bilobed appearance, measuring 138x75x110 mm in total size, located adjacent to the right side of the bladder.

The patient has also undergone a PET-CT scan, which identified a mass with irregular borders and lobulated contours measuring approximately 13x10 cm in the axial plane and up to 17 cm obliquely in the craniocaudal axis. The mass starts superiorly to the bladder in the pelvis and extends to the level of the umbilicus in the abdominal midline. The scan showed heterogeneously increased FDG metabolism, compatible with the recurrence of the primary lesion (Figure1).

Figure 1.

A Pre - Op transverse plane CT showing the mass in the abdomen



In our clinic the physical examination findings indicated presentation of asymmetry on the right side of the abdomen, hepatomegaly and an abdominal palpable mass in the right lower quadrant was dedected while palpating the abdomen.

Based on the patient's history and the results of our examinations, informed consent was obtained, and an operation was planned for the patient, who was diagnosed with a recurrent intra-abdominal mass.

2.1. Surgery

A laparotomy procedure resulted in the removal of two soft tissue tumors measuring 20 cm and 25 cm from the right lower quadrant of the abdomen. These tumors were attached to the bowel loops but showed no signs of invasion. The tumors were successfully dissected and removed from the abdomen without any major bleeding.

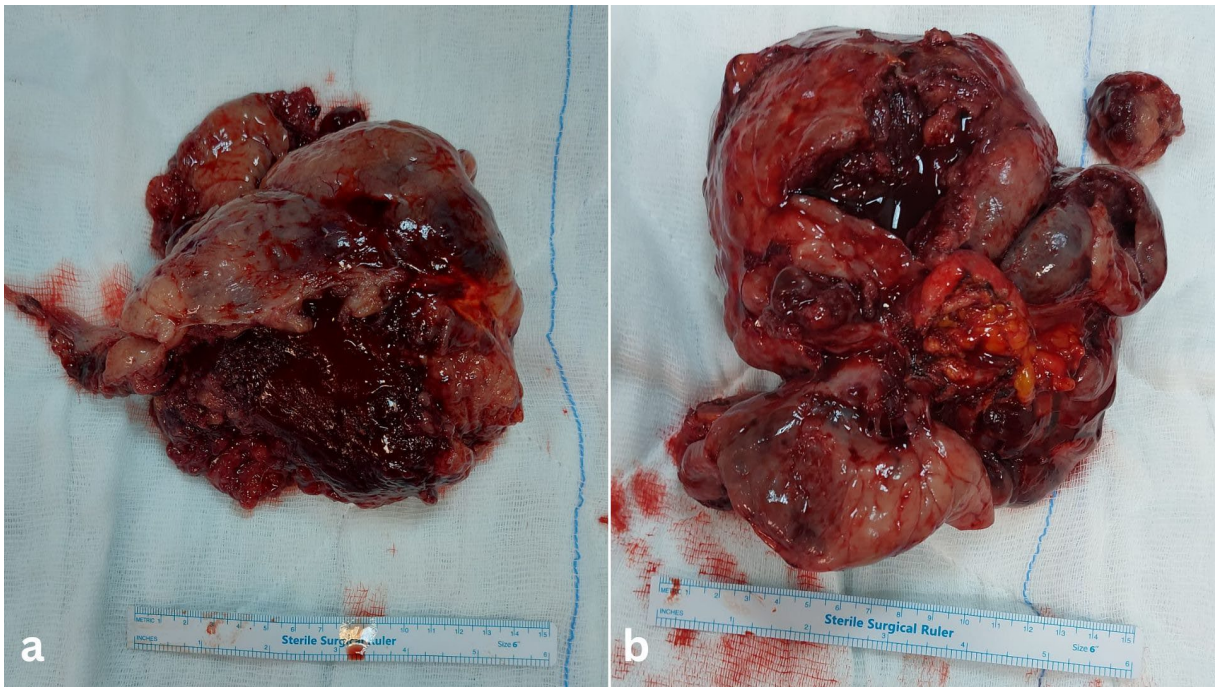
Post operative follow up was uneventful and the patient was discharged on third post operative

day. On post operative 7th day, abdominal ultrasonography revealed no fluid collection.

The post-operative pathology report confirmed the diagnosis of PEComa (perivascular epithelioid cell tumor). The tumor exhibited irregular borders and a characteristic fleshy consistency. Notably, sections of the mass showed areas of bleeding and a necrotic appearance over a wide region, indicating significant tissue breakdown and hemorrhage within the tumor. Immunohistochemical staining was performed to further characterize the tumor and confirm the diagnosis. The results were positive for HMB45, Melan A, SMA (Smooth Muscle Actin), and Desmin. These markers are commonly associated with PEComa and help distinguish it from other types of tumors. The positive staining for these markers supported the diagnosis of PEComa and provided valuable information for determining the tumor's origin and potential behavior (Figure2).

Figure 2a – b.

The tumour (from each side) after it was excised from the patient



One year after the operation, the patient experienced a second recurrence of the tumor four months post-surgery. Following this recurrence, she underwent chemotherapy at another hospital. Currently, she is receiving only anti-cancer medications.

3. DISCUSSION

PEComas, a rare group of mesenchymal tumors, present significant diagnostic and therapeutic challenges due to their diverse clinical presentations and histopathological features. According to the literature, these tumors are characterized by the expression of melanocytic and smooth muscle markers such as HMB45, Melan A, SMA, and occasionally Desmin, findings which were also observed in our patient's immunohistochemical profile.[1,2,9]

Several studies have noted that PEComas often occur in patients with tuberous sclerosis, where abnormalities in the TSC1/TSC2 genes lead to cell proliferation via activation of the mTOR pathway. There is also interest in TFE3 gene fusions and their correlation with the family of perivascular epithelioid cell tumors.[9,10,11]

Surgical resection remains the primary treatment for PEComas, as demonstrated in our patient, leading to an improvement in symptoms. Adjuvant therapies like chemotherapy have shown varied outcomes, particularly in high-risk patients. This underscores the importance of personalized treatment approaches based on factors such as tumor size, location, and molecular characteristics.[7,8]

Regarding targeted therapy, mTOR inhibitors have been shown to reduce the rate of progression in patients with metastatic disease (For Example: rapamycin or everolimus). The recurrence rate of PEComas underscores the importance of long-term surveillance and a multidisciplinary management strategy, as seen in our patient who had two reoccurrences in the span of three years.[8]

Given the rarity of PEComas, further research is essential to better understand their pathogenesis and identify new therapeutic targets. In conclusion, while advances in diagnostic techniques and treatment options have led to improved outcomes, ongoing research remains vital to enhance our understanding and management of PEComas.

4. CONCLUSİON

PEComas, a rare group of mesenchymal tumors, present significant diagnostic and therapeutic challenges because of their diverse clinical presentations and histopathological characteristics.

We report a 47-year-old female with a history of abdominal PEComa who presented to our clinic with an abdominal tumor recurrence that had increased in size. The treatment included surgical removal of the tumor and regular follow-ups to monitor for any further recurrences. Subsequent follow-ups revealed a second recurrence, after which the patient received chemotherapy at another facility.

Although advances in diagnostic techniques and treatment options have improved outcomes, continued research is crucial to deepen our understanding and improve the management of PEComas.

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No conflict of interest or common interest has been declared by authors.

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