Case Report

Pecoma of the ischiorectal fossa presenting in pregnancy - a rare presentation of a rare tumor

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Abstract

Perivascular epitheloid cell tumor (PEComa) is an uncommon entity composed of distinctive perivascular epitheloid cells with variable immuno reactivity for melanocytic and muscle markers. They are recently described tumors, and in the last decade there have been reports of similar tumors in various sites, such as uterus, retro peritoneum, breast, cardiovascular and central nervous system. However in our opinion, such a tumor has not been reported in pregnancy till now. Here we report a case of PEComa occurring in a 22 year old pregnant lady in the ischiorectal fossa, and describe the diagnostic and surgical challenges faced. We also highlight the peculiar clinical and morphological features of the case. The importance of considering PEComas in the differential diagnosis even in pregnancy, the need to arrive at a conclusive preoperative diagnosis and the requirement for immune histochemical work up have been discussed, with a revision of the concerning scientific literature.

Keywords: Ischiorectal fossa, melanocytic and muscle marker, PEComa.

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INTRODUCTION

Perivascular epitheloid cell tumors (PEComas) are characterized by their perivascular location, with a radial arrangement of cells around a vascular lumen. They are ubiquitous tumors of mesenchymal origin, described in various organs. They are composed of histologically and immunohistochemically distinctive perivascular epithelioid cells (PECs). First described in 1943 by Apitz as an abnormal myoblast in renal angiomyolipoma (AML), they were later linked to a family of tumors includingclear cell "sugar tumor" of the lung (CCST), lymphangioleiomyomatosis (LAM), clear cell myomelanocytic tumor (CCMMT) and unusual clear cell tumors of the abdominal viscera and serosa in which similar PECs were present[1,2,3]. PEComasare related to the tuberous sclerosis complex (TSC) as similar genetic alterations of TSC such as losses of TSC1 (9q34) or TSC2 (16p13.3) genes have been demonstrated in a significant number of PEComas, occurring both in TSC and in sporadic cases[2,4].

CASE PRESENTATION

A 21year old pregnant lady was first diagnosed with a mass in the ischiorectal fossa region (3 x 2 cm) picked up in a routine last trimester antenatal scan. No intervention as done at that time and advice to have follow up with the surgery department after her delivery was given. Two months after an uneventful full term normal delivery she presented to the surgical OPD with a painless swelling in the anal region. There was no history of a previous illness or surgery or any family history of cancer or familial disease. An initial ultrasonogragram (USG) revealed a cystic mass in the right ischiorectal fossa, suggesting an ovarian cyst. It was followed by a CT scan, which showed a large cystic mass lesion with enhancing intracystic solid components seen in the right side of pelvis and attached to the right inferolateral pelvic wall. The mass was seen indenting the urinary bladder and

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displacing the sigmoid colon to the left side. There were no adjacent bony erosions or destruction. (Fig 1and2) The patient underwent a USG guided true cut biopsy which yielded minimal diagnostic material due to the firmness of the swelling. Histopathological examination by Haematoxylin and Eosin stain (H and E) showed fragments of fibro collagenous tissue with extensive hyalinization. A report suggesting leiomyoma with hyalinization was given. Without a definitive pre operative diagnosis the patient was taken up for exploratory laparotomy. The surgery was complicated by severe heamorrhage from the tumor bed site. Intra operatively a markedly vascular mass at the right ischio rectal fossa confined to the infra pelvic region was encountered. Excision of the tumor was done with ligation of multiple feeding vessels, finally leading to ligation of the right internal iliac artery to rescue the patient from haemodynamic shock. Several units of whole blood and fresh frozen plasma were also transfused. The surgery was terminated with a decision to relook after 72 hours. When the packs were removed after 72 hours, oozing was again noted, which was controlled amicably. The specimen was sent for histopathological examination, the findings of which are described below. The patient underwent a prolonged hospital stay of 20 days and discharged with advice to follow up after a month. After a period of 2 months, the patient presented to the surgical outpatient department with abdominal discomfort. A USG revealed a recurrence of the tumor at the same site measuring 8 x 6 cm (almost the initial size). The patient was referred to a higher centre where a pelvic excenteration was done, followed by chemotherapy. After 38 months of follow up, she is alive with no evidence of recurrence of the tumor or any metastasis. . The patient was evaluated for tuberous sclerosis and was found negative.

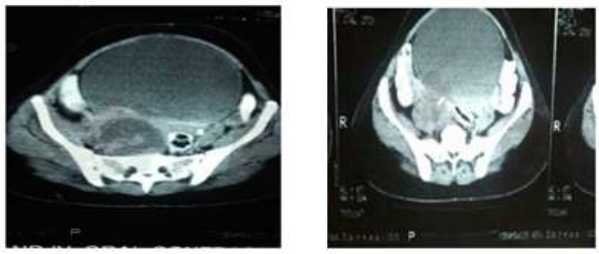

Figure 1 and 2: CT scan picture showing Large Cystic mass lesion with enhancing intracystic solid components seen in the right side of pelvis and attached to the right inferolateral pelvic wall

Figure 3 and 4: Gross picture showing firm grey tan cystic mass measuring 9x7x2 cm. External surface- irregular, grey brown and shaggy with attached skeletal muscle fibres. The inner surface was corrugated, tan brown with focal heamorrhages and spotty calcification. The cyst wall was firm and gritty at places, with thickness varying from 0.4 to 0.8 cm of the tumor

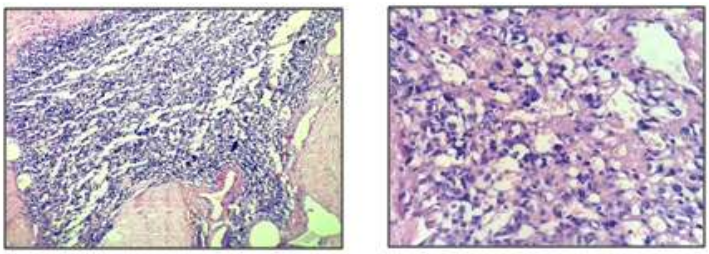

Figure 5 and 6: 10x and 40x respectively shows tumor composed sheets of uniform epithelioid cells with clear cytoplasm and vesicular nuclei, arranged around proliferating capillary sized vessels lined by plump endothelial cells. There was mild nuclear atypia and these cells were seen infiltrating the underlying dense and sclerotic connective tissue

PATHOLOGICAL EXAMINATION

The surgical specimen (excision biopsy) was fixed in 10% formalin and processed for histological examination by conventional methods. It was stained with H and E stains, reticulin and immune stained using a panel of IHC antibodies at a higher centre.

Macroscopy

Gross examination showed an already cut open firm grey tan cystic mass measuring 9x7x2 cm. External surface was irregular, grey brown and shaggy with attached skeletal muscle fibres. The inner surface was corrugated, tan brown with focal heamorrhages and spotty calcification. The cyst wall was firm and gritty at places, with thickness varying from 0.4 to 0.8 cm (Fig 3and4) Microscopy revealed a cystic tumor, the wall was composed of nodules of hyalinised connective tissue enclosing lobules, nests and sheets of uniform epithelioid cells with clear cytoplasm and vesicular nuclei, arranged around proliferating capillary sized vessels lined by plump endothelial cells. There was mild nuclear atypia and these cells were seen infiltrating the underlying dense and sclerotic connective tissue. There were foci of ectatic dilatation of vessels and hemorrhage. Occasional peripheral areas showed aggregation of tumor cells and foci of calcification. Few dispersed spindle cells were present in the intervening areas. The reticulin stain delineated the vascular channels clearly (Fig 5and6). There was minimal mitotic activity of 0-1/ 10 HPF without any foci of necrosis. IHC revealed the the following: Diffuse cytoplasmic positivity for Vimentin, S100 and HMB45 with focal positivity for Smooth muscle actin (SMA) in the tumor cells, whereas the following antibodies tested negative- Melan A, H-Caldesmon, Desmin, CD 34, CD 31, Cytokeratin, CD 68, CD74A, CD 3 and epithelial membrane antigen (EMA).We viewed this neoplasm as PEComa on the basis of morphological and immunohistochemical findings and graded it as unknown malignant potential based on the criteria put forth by Folpe *et al*[5].

DISCUSSION

PEComas are tumors of uncertain histogenesis and malignant potential that display morphologic and immunophenotypic overlap with smooth muscle neoplasia.[6] In 1996, Pea *et al* proposed the perivascular epithelioid cell (PEC) as the cell of origin for these tumors.[7] Because PECs may express both muscle and melanocytic markers, it has been hypothesized that they have the potential for melanocytic, smooth muscle, pericytic and perivascular epithelioid differentiation. Many of the reported PEComas have been associated with the tuberous sclerosis complex (TSC), an inherited disorder characterised by mutation of the TSC1 and TSC2 genes, two tumor suppressor genes located on chromosomes 9q34 and 16p13.3, respectively. This chromosomal imbalance has been demonstrated in TSC and in PEComa.[2] Ever since, there have been reports of PEComas arising in various sites, most commonly at visceral (especially uterine and gastrointestinal), retroperitoneal, and abdomino pelvic, with few occurring in soft tissue and skin with a marked female predominance.[1,8,9] This is the first documented case to our knowledge of a PEComa being reported in pregnancy. The presence of the tumor in the last trimester did not affect the course of pregnancy, and no intervention was done keeping in mind the site of the tumor and the relative symptomless nature of it. However it proved to be a locally aggressive tumor with recurrence. The inconclusive pre operative radiological and histological diagnosis led to an unexpected heavy loss of blood that required a second surgery to overcome the bleeding episode. Moreover post surgical histological diagnosis, required a panel of many antibody markers to arrive at the diagnosis. Due to the rarity of the tumor, especially unreported in pregnancy, a differential diagnosis of PEComa was not thought of. Clear criteria for malignancy in PEComas have not been elaborated, owing to the rarity of the tumors. However the WHO has put forth a number of features like infiltrative growth, hypercellularity, nuclear features, mitotic activity and necrosis have been regarded as malignant based on prior reports.[1] We classified our case, based on the criteria proposed by Folpe *et al*, which included size of the tumor as one of the defining feature. They classified these tumors as "Benign, of uncertain malignant potential and malignant"; observing a significant association between tumor size >5cm, infiltrative growth pattern, high nuclear grade, necrosis and mitotic activity >1/50 HPF and subsequent aggressive clinical behaviour of PEComas.[5]Our case was a cystic yet cellular tumor of more than 5cm; it neither showed increased mitotic activity nor necrosis. We therefore graded our case as PEComa of unknown malignant potential. The aggressiveness of the tumor was seen by the recurrence of the tumor to almost the same size, 2 months post surgery. A pelvic excenteration was done, followed by chemotherapy at a tertiary care centre. Surgery remains the mainstay for aggressive cases like our case as therapeutic trials with chemotherapy and radiotherapy are obviously difficult due to the rarity of the disease.[2] The preoperative radiological and pathological diagnosis, followed by the post operative histological diagnosis and the surgical management were all challenges in their own way. We need to consider PEComas in the differential diagnosis of both soft tissue and cystic tumors, especially in females albeit pregnancy. We also require constant

long term follow up and treatment modalities of more number of such cases to arrive at a common consensus to deal with these rare tumors which have proved to be an enigma both to the pathologists, radiologists and the clinicians.

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