Malignant Subcutaneous Perivascular Epithelioid Cell Tumour of Anterior Abdominal Wall

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ABSTRACT

Perivascular epithelioid cell tumours (PEComas) are a family of rare mesenchymal tumours with discrete histologic and immunohistochemical characteristics. Even rarer among them are cutaneous and subcutaneous PEComas. We describe a 34-year-old lady presented with a large anterior abdominal subcutaneous lesion showing intact overlying skin and no obvious invasion of abdominal musculature. A wide local excision was performed. Histopathology revealed a solitary tumour measuring 75 x 55 x 90 mm with epithelioid cells in nests with thin fibrovascular septa and spindle cells. Resection margins were clear with no invasion to skin or rectus sheath. Tumour cells were only positive for HMB-45 but negative for other markers. This is the largest subcutaneous PEComas reported to date.

Keywords: Clear-Cell Sugar Tumours, PEComas, Perivascular Epithelioid Cell Neoplasms

INTRODUCTION
Perivascular epithelioid cell tumours (PEComas) are a family of rare mesenchymal tumours closely related to angiomyolipomas, lymphangioleiomyomatosis and clear cell sugar tumour of the lung. They have no known benign cell counterpart, but are made up of perivascular epithelioid cells displaying both melanocytic and smooth muscle differentiation; with specific histologic and immunological features.\(^1\) PEComas have been described in the literature originating from various sites such as pelvis, genital tract, lung, as well as intra-abdominal organs; with cutaneous/subcutaneous PEComas exceptionally rare among these tumors.\(^2\) There is a paucity of literature describing PEComas over the abdominal wall. Here we present a rare case of malignant subcutaneous PEComas over the abdomen, the first ever described in Southeast Asia.

**CASE REPORT**

A 34-year-old healthy lady presented with a rapidly enlarging anterior abdominal wall mass noticed over the duration of 3 months. She was otherwise well apart from occasional pain over the lesion. There was no family history of similar conditions. Physical examination revealed a large subcutaneous mass that was firm, mobile with increased prominence upon muscle contraction. Overlying skin was normal. An ultrasound showed a solid lobulated mass with vascularity indenting the abdominal wall. A trucut biopsy demonstrated an inflamed fibrocollagenous tissue infiltrated by epithelioid tumour cells. A computed tomography (CT) scan of the abdomen showed a heterogenous lobulated subcutaneous lesion at the left anterior abdominal wall measuring 54 x 73 x 68 mm with central necrosis [Figure 1A and 1B]. There was no calcification. There was poor plane of demarcation with the abdominal muscles but no evidence of distant metastasis.

She was decided for a wide local excision, in which it was performed under general anaesthesia as she has an acceptable anaesthetic risk. An elliptical incision was made on a previously made skin marking [Figure 2A]. Intraoperatively, there was a mobile tumour measuring 8 x 6 cm with the posterior part attaching to the anterior rectus sheath. A wide local excision with 1 cm margin was performed, resecting along the involved anterior rectus sheath. The feeding vessel is from superior epigastric artery and it was carefully identified, doubly ligated and divided. A non-degradable polypropylene mesh was incorporated on the breached segment to avoid future herniation [Figure 2B]. A vacuum drain was inserted to avoid risk of deep surgical site infection from haematoma collection.
The resected specimen revealed a solitary tumour measuring 75 x 55 x 90 mm [Figure 2C & 2D]. The cut section of the tumour showed a friable with golden yellow tan orange cut surface and area of haemorrhage. All resection margins were tumour-free with the closest margin of 5 mm at the deep margin. Histology demonstrated epithelioid cells in nests with rounded nuclei, irregular nuclear membrane and abundant eosinophilic to clear cytoplasm with thin fibrovascular septa and some spindle cells with evidence of lymphovascular invasion [Figure 3A-3C]. It showed moderate to marked nuclear atypia and nuclear pleomorphism with many large and bizarre nuclei. Less than 50% of coagulative necrosis was observed with mitotic figures of 3 in 10 HPF. Immunohistochemistry (IHC) revealed that the tumour cells were positive for Human Melanoma Black (HMB)-45 [Figure 3D] but negative for desmin [Figure 4A], myogenin, Melan A [Figure 4B], PanCKAE1/AE3 [Figure 4C], U79 RCC marker [Figure 4D], S100, vimentin, SMA and CD10. This tumour was graded as the French Federation of Cancer Centers Sarcoma Group (FNCLCC) Grade 2. She was discharged at day 3 post operatively and completed 50 Gy/25 fractions (2 Gy per fraction) of adjuvant radiotherapy. She is currently well with no signs of recurrence.

**DISCUSSION**

Most cases of PEComas have been reported to occur in females with a median age of 43 years old which shows a strong female predominance despite excluding those of gonadal origin. As mentioned above, cutaneous/subcutaneous PEComas are rare, with only 48 cases reported till present. Of these, only three cases including ours are completely subcutaneous with no dermal involvement.\(^1,3,4,5\) Most of these tumours measure around 10-20 mm, making ours by far the largest to date.

Cutaneous PEComas characteristically present in a well-demarcated dermal lesion which is composed of epithelioid cells with a large, clear or slightly granular cytoplasm and nuclei arranged in a nested or trabecular pattern intermingled with spindle cells. These cells are arranged within fine vasculature ranging from capillaries to thicker arterioles and arteries.\(^6\) Various cellularities are described with low to moderate cellularity the commonest of all. Several cases have also described multinucleated giant cells with a central zone of eosinophilia which is not found in our case. An important histological finding to note is the
mitotic activity which can range from 0 to 50 mitoses/50 HPF as atypical mitotic figures are only present in a minority of cases. Some tumours have coagulative cell necrosis which may be representative of the larger sized tumours.  

The characteristic IHC of PEComas has contributed to the diagnosis of soft tissue tumours which expresses at least 1 melanocytic marker. They most commonly are positive for HMB-45 as in our case and sometimes are positive for Melan-A, tyrosinase and microphthalmia transcription factor. Approximately half display positivity for muscle markers such as actin, desmin, vimentin, myoglobin and myosin. Calder et al described the negativity of S-100 in PEComas to differentiate between melanoma however 33% of cases were said to be positive to S-100 in a case series reported by Folpe et al. To date, there has only been 3 cases report describing a negative expression of PEComas to HMB-45.

The majority of cutaneous PEComas are located at the extremities and display benign features. However, it appears that those on the scalp, face and trunk (as with ours) are more likely to display malignant features. Folpe et al proposed a classification of PEComas to define the criteria of malignancy in 2005 and classified PEComas into either “benign”, “uncertain malignant potential” or “malignant”. This was derived from the strong association of several features to recurrences and metastasis which includes tumour size of more than 5 cm, infiltrative growth pattern, high nuclear grade, necrosis and mitotic activity of >1/50 hpf. It was then built on by Bleeker et al again in 2011 for risk stratification focusing on the risk of recurrence and they found that the size of >5cm and high mitotic rate were the only two factors significantly associated with recurrence after surgical resection. Our case shows a strong tendency to recur in view the aforementioned risk namely size and mitotic rate, hence an adjuvant radiotherapy was provided and a vigorous follow up is required to ensure that she will be in remission.

CONCLUSION

PEComas are a rare entity with variable presentation and tumour behaviour. It is important to always consider rarities when confronted with an atypical skin lesion. A multidisciplinary preoperative evaluation should include information obtained by fine needle or core biopsy so that the appropriate surgical approach can be planned.
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REFERENCES


Figure 1: (A) CT scan of the abdomen showed a heterogenous lobulated subcutaneous lesion (white arrow) at the left anterior abdominal wall in axial view. (B) The lesion (white arrow) at another angle in sagittal view.
Figure 2: (A) Preoperative marking depicting the mass and incision. (B) Mesh repair of the rectus sheath defect post excision of the tumour. (C) Posterior surface of the resected specimen, showing the rectus sheath resected along with the tumour. (D) Side view of the resected specimen.
Figure 3: (A) H&E staining showing epithelioid cells in nests with thin fibrovascular septae (black arrow). (B) H&E staining showing epithelioid cells (white arrow). (C) H&E staining showing lymphovascular invasion (black arrow). (D) Epithelioid cells with positive staining with HMB-45.
Figure 4: Negative IHC of desmin (A), Melan A (B), PanCKAE1/AE3 (C), U79 RCC marker (D).