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Case Report

Uterine Perivascular Epithelioid Cell Tumour Presenting as a Cervical Mass

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Abstract

Perivascular Epithelioid Cell Tumour (PEComa) also known as myelomelanocytic tumours are uncommon, recently described mesenchymal tumours that include angiomyolipoma, clear cell "sugar" tumour of the lung, lymphangioleiomyoma and tumours composed predominantly of epithelioid cell morphology. A predilection for uterus has been described. However up till now only 14 cases of uterine PEComas have been described. All of these were seen in adult females in the peri and post menopausal age group (from 40-75years), and almost all were located in the region of body of uterus. A single case in the upper cervical region has been reported with uterine PEComatoses. We report an unusual presentation of this rare tumour presenting as a polypoidal cervical mass in a young female. Occurrence of this tumor in a young female as seen in our case warrants inclusion of PEComa in the diferential diagnosis of all epithelioid and clear cell neoplasms of uterus irrespective of age.

Introduction

Perivascular Epithelioid Cell Tumour (PEComa) also known as myelomelanocytic tumours are uncommon, recently described mesenchymal tumours that include angiomyolipoma, clear cell "sugar" tumour of the lung, lymphangioleiomyoma and tumours composed predominantly of epithelioid cell morphology.[1] A predilection for uterus has been described. However, up till now only 14 cases of uterine PEComas have been described. All of these were seen in adult females in the peri and post menopausal age group (from 40-75years), and almost all were located in the region of body of uterus.[2] A single case in the upper cervical region has been reported with uterine PEComatoses.[3] We report an unusual presentation of this rare tumour presenting as a polypoidal cervical mass in a young female.

Case Report

The patient was a 25 year old multiparous lady from northern areas of Pakistan, who had presented with irregular vaginal bleeding. The cervical examination revealed a fungating growth about 4cm on the anterior lip of the cervix. The biopsy of that lesion had revealed adenocarcinoma and she was referred to the tertiary care centre in Karachi i.e., Aga Khan University Hospital. In our institution, she underwent a formal staging procedure by examination under anaesthesia and cystoscopy which revealed stage 1b1 disease. An MRI abdomen and pelvis correlated well with the clinical staging. She finally underwent radical hysterectomy and pelvic lymphadenectomy. Her post-operative recovery was satisfactory.

The resected hysterectomy specimen revealed an ulcerated polypoidal lesion in the ectocervix measuring 3.5x3.0x1.5cm. The submitted sections showed a clear cell neoplasm with an alveolar growth pattern. Surface ulceration was seen, however no dysplastic changes were present in the intact epithelium. The tumour cells exhibited varying degree of cytoplasmic clearing with some cells showing eosinophilia. The nuclei were mildly pleomorphic with occasional prominent nucleoli. An infiltrating pattern of growth was observed with tongues of tumour extending into the surrounding stroma. However the tumour was confined to the cervix with no evidence of involvement of endometrium or adnexae. On periodic Schiff (PAS) stains the tumor cells were strongly positive for glycogen whereas reticulin stains showed prominent septa but was not surrounding individual cells. On immunohistochemistry, tumour cells were positive for vimentin and HMB 45 and negative for cytokeratins, epithelial membrane antigen, desmin, Alpha Smooth Muscle Actin (ASMA) and S-100. All recovered pelvic lymph nodes were tumor free.

Discussion

Uterine PEComas are uncommon tumours, described initially by Bonetti, composed at least in part of perivascular epithelioid cells.[1] Histologically the cells have abundant clear to eosinophilic cytoplasm and are distinguished from epitheliod smooth muscle tumors by expression of HMB45 with a variable expression of ASMA. Molecular studies fail to demonstrate any significant chromosomal aberrations and most tumours are found to have a diploid DNA.[6] As only a small number of cases have been reported so far, the prognosis of these tumours is not well defined. Of the 13 cases of uterine PEComa described so far, 4 have behaved in an aggressive manner. Three cases were locally aggressive with recurrence and extension into adnexa whereas a single case showed metastases to lung after a period of 7 years.

At least some of the cases are associated with tuberous sclerosis complex.[4] No such association, however, was seen in our case. Two morphological patterns have been described. One composed of cells with abundant clear to eosinophilic granular cytoplasm, diffuse positivity for HMB45 with focal expression of smooth muscle markers and a tongue like growth pattern. The other group is composed of epithelioid cells with less prominent clear cell features and focal expression of HMB45.[5] Our case falls in the first group except for negativity for smooth muscle actin.

As only a small number of cases have been reported so far and the clinical presentation, microscopic appearance and clinical behavior differs widely for each case, PEComas are at best considered as tumours of uncertain malignant potential. Most cases reported in the literature were seen in late middle and old age. Occurrence of this tumour in a young female as seen in our case warrants inclusion of PEComa in the differential diagnosis of all epithelioid and clear cell neoplasms of uterus irrespective of age.

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