

Diagnostic pitfalls of pilomatricoma on fine needle aspiration cytology

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Pilomatricoma is a benign skin adnexal tumour usually seen in the head and neck region of children and young adults. It is underrecognized on cytology, resulting in the overdiagnosis of malignancy. We bring forth a case report of a slow growing nodular swelling in a 10-year-old female child, which was misdiagnosed on Fine Needle Aspiration Cytology (FNAC) as a malignant neoplasm and found to be pilomatricoma histopathologically. Nodular lesions of pilomatricoma, which are predominantly composed of basaloid cells and devoid of other diagnostic features, lead to a false diagnosis of malignancy. To avoid misdiagnosis, pilomatricoma should be considered in differential diagnosis when primitive appearing cells are aspirated in slow growing subcutaneous nodules. Cytomorphological features of pilomatricoma and major pitfalls in diagnosis are also being discussed.

Keywords: adnexal tumour, fine needle aspiration cytology, pilomatricoma

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INTRODUCTION

Pilomatricoma, also called calcifying epithelioma of Malherbe, is a benign skin adnexal tumour of hair matrix origin. It accounts for 20% of all hair follicle related tumours and usually occurs in the head and neck region. It presents as a solitary, slow growing dermal or subcutaneous nodule and is rarely diagnosed clinically^{1,2}. Histopathologic features of this lesion are characteristic and well recognized but diagnosis is sometimes difficult on cytology³. It is repeatedly misdiagnosed cytologically as an epidermal inclusion cyst, giant cell tumour, squamous cell carcinoma, malignant adnexal tumour or metastatic neoplasm⁴⁻⁸.

CASE REPORT

The patient was a 10-year-old female child who presented with a left cervical swelling of 1.0×1.0cm for six months with multiple episodes of pain. A clinical diagnosis of left cervical lymphadenopathy

was made. On ultrasonography, multiple enlarged lymph nodes were seen along with a hypochoic lesion measuring approximately 2.0×1.5 cm with foci of calcification in the left submandibular region. FNA was requested. On local examination, a firm to hard freely mobile nodule measuring 1.5×1.3 cm was seen. FNAC was performed using 23 Gauge needle and smears were stained and examined. On microscopic examination, the smears were moderately cellular and showed atypical cells (non lymphoid) having round to oval nuclei with mild pleomorphism, reticulogranular chromatin and scanty to moderate amount of cytoplasm which was granular at places. (Figure 1) Accordingly, the diagnosis of a neoplastic lesion possibly malignant was made and excision biopsy was advised.

On further examination, all her systems were normal, investigations including hematological parameters, chest radiographs and abdominal ultrasound were normal. The cervical nodule measuring 2.0×1.5×1.0 cm was excised. The cut surface of the nodule was grey white and chalky.

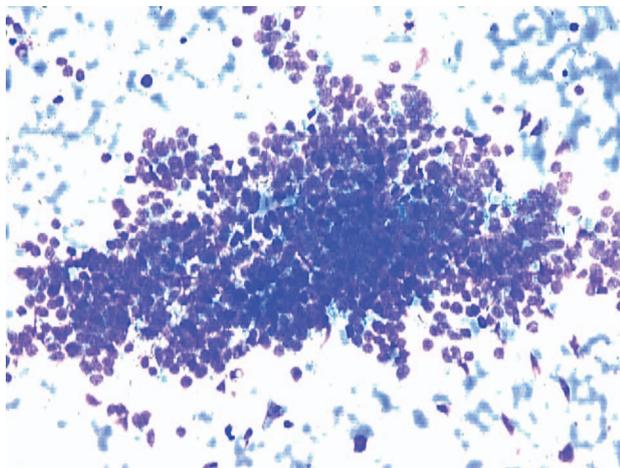


Figure 1. Photomicrograph showing mainly basaloid cells. (Giemsa*10)

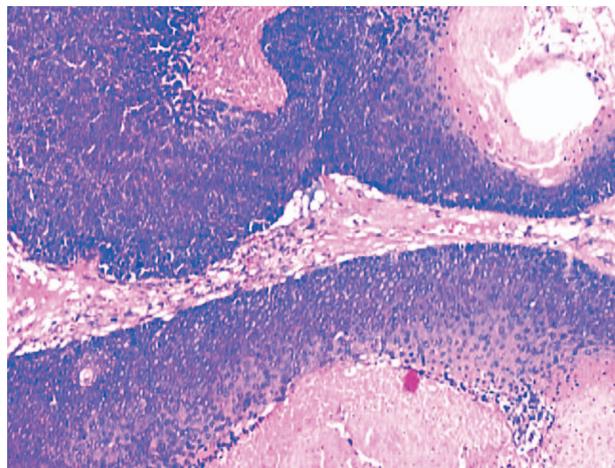


Figure 3. Photomicrograph of pilomatricoma showing basaloid cells and ghost cells. (H&E *10)

(Figure 2) Histopathological examination of the excised specimen showed typical histology of pilomatricoma, comprising mainly nests of basaloid cells, few islands of ghost cells along with many foreign body giant cells and areas of calcification. (Figure 3)



Figure 2. Photograph of gross specimen of pilomatricoma.

DISCUSSION

Pilomatricoma is a benign cutaneous appendageal tumour with differentiation towards the matrix and inner sheath of the normal hair follicle and cortex. It occurs in hair bearing areas with a predilection for the head and neck region, the other sites being upper extremities, trunk and lower extremities⁹. Clinically, it presents as a solitary slow growing dermal or subcutaneous nodule commonly seen in children and young adults with age of presentation showing a bimodal pattern with the first peak being 5-15 years and the second being 50-65 years, with a female preponderance¹⁰.

Patients usually present with a solitary nodule, growing slowly over several months or years. Most of the time, patients are asymptomatic with a few complaining of pain. The lesions range from 0.5-3.0 cm but rarely, lesion as large as 5 cm have been reported¹. On palpation, they are single, firm and stony hard nodules. Plain X-rays reveal nonspecific calcification while ultrasonography shows a well defined round hyperechoic mass with post dense acoustic shadows. Computed tomography and magnetic resonance imaging reveal a sharply demarcated subcutaneous opaque lesion that does not enhance on injection of contrast media or small areas of single dropout consistent with the presence of calcification¹¹. The cytologic diagnosis is based on combination of basaloid cells, ghost cells and foreign body giant cells against an inflammatory background. Basaloid cells have high Nucleus: Cytoplasm (N: C) ratio and can be seen as single cells or in clusters, sheets or as bare nuclei in the

background while ghost cells are usually seen in clusters¹². Despite well described features, and FNAC been performed as a diagnostic tool, the lesions predominantly composed of basaloid cells have repeatedly been misinterpreted as malignancy, primary or metastatic, because of their high N:C ratio, mild nuclear hyperchromasia and the presence of nucleoli. On the other end of the spectrum, when ghost cells or foreign body giant cell predominate, the cytologic features can even mimic epidermal inclusion cysts or giant cell lesions⁷. In our case, the possibility of neoplasm was considered as the lesion was showing predominant population of basaloid cells. However, patient's age, clinical history of six months duration and lack of significant nuclear pleomorphism resisted us to definitely conclude the possibility of malignancy and surgical biopsy was advised. It is to be further added that all the diagnostic features may not necessarily be present in every case, especially when the aspirate is from the periphery of the lesion. In such cases, the spectrum of characteristic cellular components and predominance of one component over the other leading to the diagnostic trap should always be considered and help the pathologist to avoid incorrect diagnosis on cytology.

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