CASE REPORT

Diagnostic Pitfall in Fine Needle Aspiration Cytology of Pilomatrixoma with Unusual Clinical Presentation

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ABSTRACT

Pilomatrixoma (PMX) is a benign skin adnexal tumour with matrical differentiation. It frequently presents as a painless and slow growing solitary skin nodule primarily at the head, face and neck regions. Although there is increasing understanding on the clinical presentations and morphological features of PMX, difficulties are still expected in establishing the clinical and cytological diagnosis. We report a young girl who presented with a painless post-auricular swelling for one year with sudden increased in size. Computed Tomography (CT) scan and fine needle aspiration cytology (FNAC) findings were suggestive of a malignancy. Diagnosis of PMX was established and confirmed by tissue histopathological examination. The purpose of this study is to demonstrate the diagnostic pitfall of PMX in FNAC specimens, especially in patients with unusual clinical presentations.

Keywords: Pilomatrixoma, Skin nodule, Fine needle aspiration cytology

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INTRODUCTION

Pilomatrixoma (PMX) which is also known as 'calcifying epithelioma of Malherbe', shows differentiation towards hair matrix portion of the lower segment (stem) of hair follicles. It is more commonly encountered in young age group with slight female predilection. PMX frequently occurs at the head, face and neck as well as the upper extremities. PMX nodules are usually asymptomatic. However, in deeply-seated nodules, stretching of the skin over the tumour may cause pain or discomfort and show a 'tent sign' with multiple facets and angles. Most lesions were small measuring less than 3 cm, but giant lesions measuring more than 15 cm have also been reported (1). Smears from FNAC are helpful in establishing an accurate diagnosis of PMX. However, due to heterogeneity of the lesions especially in cases with unusual or aggressive clinical presentations, the results of FNAC can sometimes be misleading. A spontaneous regression of PMX has never been reported. Complete surgical excision with adequate margin is the recommended treatment and recurrence after surgery is rare if the lesion is completely excised. Malignant transformation of PMX to a pilomatrical carcinoma even though rare, should be suspected in cases with repeated local recurrences.

CASE REPORT

A 7-year-old Malay girl, with no known medical illness presented with right postauricular nodule for one year. The nodule was painless and initially was gradually increasing in size. There was no history of fever, trauma, or infection. Her parents noticed sudden increase in nodular size which was associated with redness of the overlying skin one month prior to medical consultation.

On examination, the nodule measured 6 x 5 cm, non-tender, firm and slightly mobile with mild erythema of the overlying skin. There was no discharge or enlarged cervical lymph nodes on palpation. Full blood picture and blood chemistries were within normal limits. Ultrasound was suggestive of infected necrotic postauricular lymph node. The head and neck CT scan showed a well-defined enhancing heterogeneous mass with calcification (Figure 1). The impression of the CT scan finding was soft tissue sarcoma or lymphoma. Together with clinical behaviour of the lesion which was large and increasing in size, the provisional clinical diagnosis at that time was consistent with the CT scan findings.

Smears from FNAC of the right postauricular nodule demonstrated several ductal epithelial cell clusters with predominant squamoid and occasional basaloid looking cells. In some clusters, the cells exhibited overlapping enlarged vesicular nuclei with coarse chromatin pattern and occasional prominent nucleoli (Figure 2). Mitotic

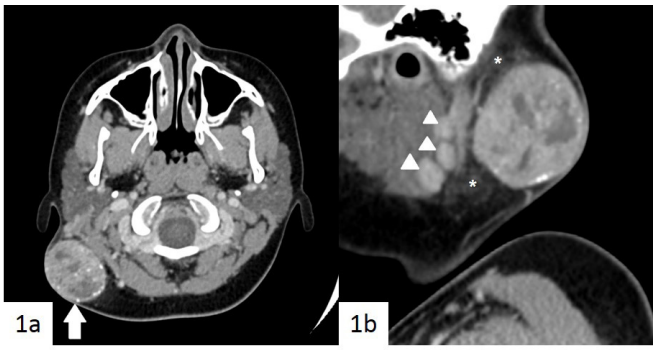

Figure 1: Axial CT scan (a) showed a well-defined heterogenously enhancing lesion at right subcutaneous retroauricular region with specs of calcification within and surrounding the lesion (arrow). On sagittal view (b), presence of streakiness of the adjacent fat (*) with enlargement of the cervical lymph nodes chains (arrowhead).

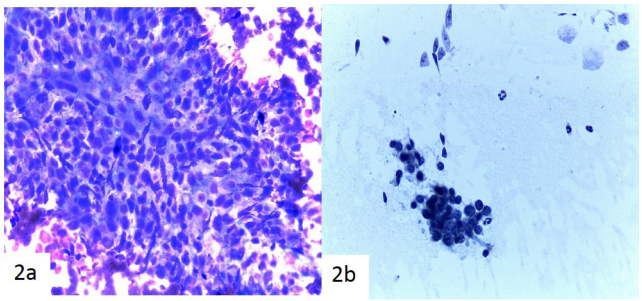

Figure 2: May-Grunwald-Giemsa (MGG) stain x 400 (a) showed cluster of tumour cell with dual cell populations of pleomorphic basaloid and squamoid-looking cells. Papanicolaou (PAP) stain x 400 (b) showed compact cluster with some overlapping basaloid cells having enlarged vesicular nuclei with coarse chromatin with some exhibit prominent nucleoli.

figures were identified. The FNAC interpretation was suspicious of malignancy, suggestive of malignant squamo-proliferative neoplasm. Excisional biopsy with adequate surgical margin was subsequently carried out and histopathological examination revealed features consistent with PMX (Figure 3). The patient was cured with no recurrence reported.

DISCUSSION

Cytological diagnosis of PMX is made based on triad features, which are basaloid cells, ghost cells and foreign-body multinucleated giant cells in background of inflammatory cells. However, this triad does not necessarily present in all cases, as it also depends on the site of the fine needle aspiration. Aspiration that is performed on an early lesion or at the periphery of the lesion will show basaloid cells predominance without other components, whereas aspirates from an older lesion may demonstrate sole presence of ghost cells (2).

The basaloid cells can be observed as dispersed single cells, sheets, clusters or bare nuclei in the background. Ghost cells on the other hand, are usually seen in clusters. The basaloid cells can be easily

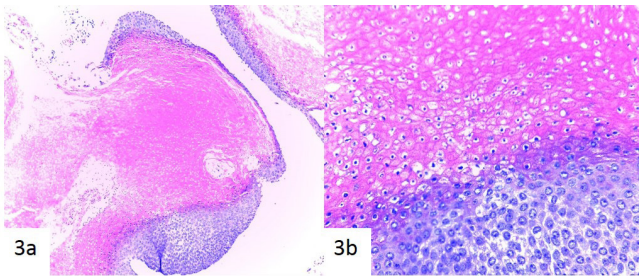

Figure 3: Histological findings showed proliferation of basaloid cells with some abrupt keratinisation and anucleated ghost cells. Haematoxylin and eosin stain, original magnification x 100 (a) and x 400 (b).

misinterpreted as basaloid squamous cell carcinoma or basal cell carcinoma (BCC) because of their nuclear pleomorphism, nuclear hyperchromatism, high nucleus:cytoplasmic ratio and large nucleoli (3). Apart from that, basaloid cells may mimic small round blue cells found in embryonal rhabdomyosarcoma, Merkel cell neuroendocrine carcinoma of the skin and metastatic small cell carcinoma of the lung (2,3,4). The basaloid cells with hyperchromatic nuclei in cohesive groups are found in other skin appendageal tumours, such as spiradenoma, adenoid cystic carcinoma (AdCC) and cylindroma. However, the absence of ghost cells, multinucleated giant cells as well as acellular calcific deposits in these conditions are crucial features in ruling out PMX (5).

In our case, the age and gender were generally consistent with PMX. However, the behaviour of the lesion, imaging and FNAC findings indicated otherwise. The ultrasound findings of PMX usually appear as a well circumscribed, non-homogeneous echogenic nodule. The key finding is early calcification of the lesion. In this case, the ultrasound findings showed a well-defined mixed solid-cystic lesion seen at the subcutaneous plane of the right posterior auricular region with no calcification identified. On the other hand, the CT scan with contrast showed a well-defined enhancing heterogenous hyperdense lesion associated with specks of calcification and multiple enlarged lymph nodes.

The FNAC interpretation was suspicious of malignancy as the smears showed clusters of ductal epithelial cells with irregular nuclear membrane, pleomorphic enlarged nuclei, prominent nucleoli, scanty cytoplasm and occasional mitoses. The cell block showed a few clusters of atypical cells with large nuclei, coarse chromatin, irregular nuclear outline, prominent nucleoli, and abundant cytoplasm.

Histopathologic examination of excised mass demonstrated the presence of basaloid cells, ghost cells and foreign-body multinucleated giant cells in the background of chronic inflammatory cells triad which were straightforward features for diagnosing PMX.

CONCLUSION

FNAC is an important pre-operative diagnostic investigation for PMX but pitfalls can arise when one component of PMX is predominant over the others. Such pitfall should always be considered to avoid a misdiagnosis as well as an overdiagnosis. Thus, performing an excisional biopsy for histopathological confirmation is recommended to establish the diagnosis in inconclusive FNAC finding. This case report emphasises the importance of cyto-histological correlation, especially in difficult cases and it should be practised by pathologists to help in delivering proper patient management.

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