Pilomatricoma of cheek mimicking malignancy: A diagnostic challenge on Fine needle aspiration cytology

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Received - 21 September 2019 Initial Review - 07 October 2019 Accepted - 12 October 2019

ABSTRACT

Pilomatricoma is a benign adnexal lesion which is often diagnosed on cytology. Sometimes cytomorphology may pose diagnostic challenge and may mimic malignant neoplasm. We present a case of a slow growing nodular swelling on the right upper cheek in a 9-year-old male child with similar diagnostic difficulty on Fine Needle Aspiration Cytology (FNAC).

Keywords: *Adnexal neoplasm, Face, FNAC, Subcutaneous swelling, SRCT.*

Pilomatricoma is an unusual, slow growing benign adnexal tumor presenting in the head and neck region and upper extremities [1-2]. This entity is also described as calcifying epithelioma [2], it is a sub-epidermal tumor and arises from protoepithelial cells or hair matrix [3-4]. It is slightly more common in females and usually noted in children in the 1st two decades of life [4-5]. Cytomorphological features are well described; however, case reports with overdiagnosis of malignancy are documented in literature [3-5]. Here, we report a case of pilomatricoma cheek in a male child with challenges in cytomorphological diagnosis.

CASE REPORT

A 9-year-old male child presented to the hospital with swelling over right cheek for last 15-20 days. He did not receive any prior treatment for this swelling. On general examination, there was no associated fever or any other complaint or any abnormality. The patient gave history of trauma with pencil at the same site 1 month back. On examination, the swelling was subcutaneous, measured approximately 1x1 cm and was firm, slightly fixed & non-tender. The overlying skin showed mild bluish discoloration [Fig. 1]. Complete blood count showed haemoglobin-13.4gm/dl, total leukocyte count of 7000/µl, platelet- 3.4 lakhs/µl. Rest other parameters including the biochemical parameters like electrolytes, thyroid hormone, liver and renal function tests were within normal limits.

A clinical diagnosis of sebaceous cyst was made and fine needle aspiration (FNA) was done from the cheek swelling. Multiple smears were prepared followed by air drying and fixing in 95% ethyl alcohol. These were subsequently stained with Giemsa and Papanicolaou stains respectively. Smears were highly cellular revealing two distinct cell population. Epithelial cells showed mild to moderate pleomorphism and were arranged in sheets, clusters, groups, singly scattered and vague gland like pattern [Fig. 2a-c]. These cells were medium sized, round to oval, having scant amount of cytoplasm, round to oval nuclei, finely dispersed chromatin, prominent nucleoli [Fig. 2d]. Also noted were elongated spindled and plasmacytoid looking cells with granular chromatin and moderate amount of pale to dense cytoplasm which are seen singly scatteredand also embedded in pink matrix [Fig. 2a & 2c]. The background showed areas of pink matrix and occasional cystic macrophages admixed with blood.

Further, cell block was also prepared, and Hematoxylin & Eosin stained section showed cluster of similar small round cells. In view of clinical details provided, cytologic possibility of adnexal tumor and small round cell tumor (SRCT) was considered. Immunohistochemistry using LCA, CD99 and MyoD1 was done. All were found to be negative in the index case.

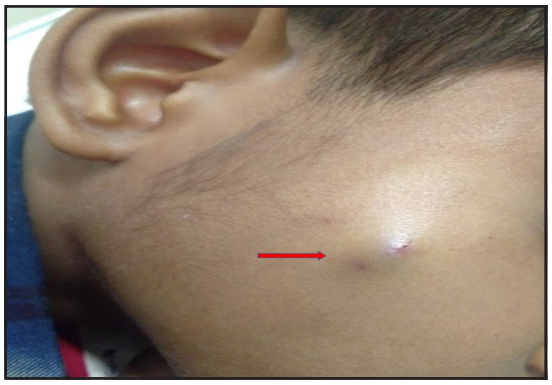

Figure 1: A subcutaneous, slightly fixed, non-tender swelling measuring approximately 1x1 cm; located on upper cheek and showing mild bluish discoloration.

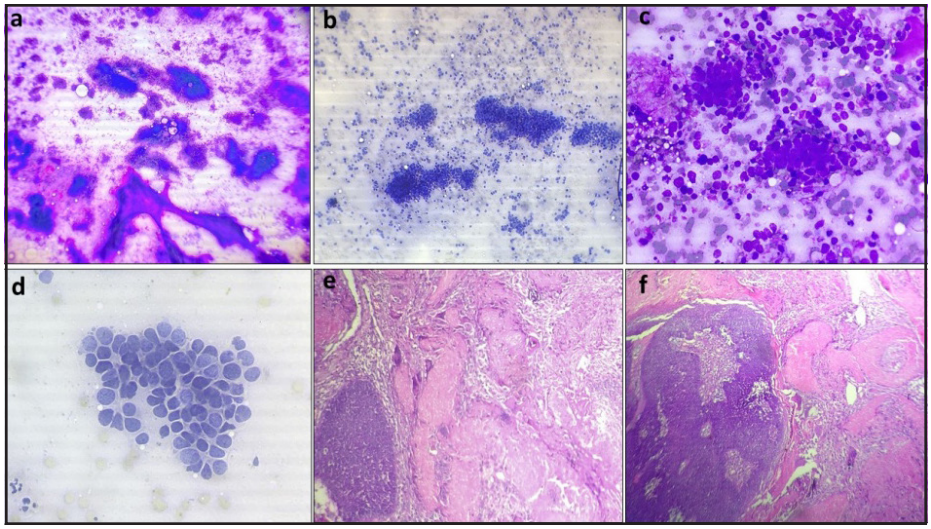

Figure 2: a-c: Cellular smears showing epithelial cells arranged in sheets, clusters, groups, papillaroid fragments, singly scattered & in vague gland-like pattern [Figure 2a- 100X, Giemsa & 2b-100X, Pap], embedded in pink matrix material [2c- 200X, Giemsa];d- Cells are medium sized, round to oval, having scant amount of cytoplasm, round to oval nuclei, finely dispersed chromatin, prominent nucleoli [Figure 2d- 400X, Pap]; e-f: Section showing many irregular large islands of basophilic epithelial cells with scant cytoplasm showing in places abrupt and gradual transition into shadow cells with occasional giant cells [Figure- e&f H&E, 200X], confirming diagnosis of pilomatricoma.

As per the advised radiological findings, contrast enhanced computed tomography (CECT) head showed an evidence of rounded, subcutaneous, non-enhancing, nodular soft tissue lesion measuring 10x8 mm. The swelling was located over the right zygomatic arch. Keeping in view the history of trauma at the same site, a provisional diagnosis of post-traumatic infective nodule was made.

In view of the radiological and clinical details, FNA slides were reviewed again and final diagnosis of an adnexal neoplasm was suggested. Surgical resection of the lesion with adherent skin was done and sent for histopathological examination. Section examined showed many irregular large islands of epithelial cells. These were masses of basophilic cells with scant cytoplasm, showing in places abrupt and gradual transition into shadow cells with occasional giant cells [Fig. 2e-f]. The histopathological diagnosis given was pilomatricoma. He was given antibiotics for a week and the recovery of the child post-surgery was good. The child was re-examined after 1 month of surgical removal of the lesion and no swelling was seen.

DISCUSSION

Pilomatricoma, also known as calcifying epithelioma of Malherbe, is a benign cutaneous tumor arising in the dermal layer of hair-bearing skin [2-3]. Clinically it presents as a solitary slow-growing, firm and mobile mass usually ranging in size from 0.5 to 5 cm. The overlying skin may take a red or blue hue. It is usually noted in the head and neck region [3]. It is commonly seen in children and young adults in the 1st two decades of life [3-4]. Pilomatricoma may be seen in association with other disorders like Gardner syndrome, Rubinstein-Taybi syndrome, Turner's syndrome, etc. [4-5].

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 calcification [5-7]. FNAC of pilomatricoma is characterized by the presence of basaloid cells, ghost (shadow) cells, many foreign body giant cells, nucleated squamous cells in the background of amorphous debris. Amongst these, ghost cells are the key diagnostic component when seen with other findings. Early lesions are predominantly composed of basaloid cells, leading to a basaloid rich aspirate. The characteristic shadow cells may be absent in such basaloid rich smear or may be overlooked due to lack of nuclei or poor staining properties [3-5].

Histopathologic features of this lesion are characteristic and well recognized, but diagnosis is sometimes difficult on cytology. Cytological differentials may be varying depending upon the combination of the cytological features obtained on FNAC which include epidermal inclusion cyst (EIC), giant cell tumour, SRCT, squamous cell carcinoma, malignant adnexal tumour or metastatic neoplasm.

The index case was predominantly composed of primitive looking basaloid cells and no other component was aspiratedon FNAC. A false positive diagnosis of SRCT (Primitive neuroectodermal tumor/ rhabdomyosarcoma) was also considered in first place. However, clinical details, lack of significant radiological findings and negative immunostains resisted us to conclude the possibility of SRCT.

Studies show that pilomatricoma has been often misdiagnosed as carcinoma [3-6]. A study by Wang et al [7] reported a high rate of incorrect diagnosis of pilomatricoma (45%). Thapliyal et al [8] also noted a case of pilomatricoma in a 32-year-old male which mimicked as Merkel cell carcinoma/ small round cell tumor on FNAC. Hence, it is well established that, despite knowing the diagnostic morphological details of pilomatricoma, it is still a diagnostic challenge; both clinically as well as cytologically.

CONCLUSION

Diagnosis of pilomatricoma on cytology may be challenging and may lead to false diagnosis of malignancy; especially in cases where basaloid cells predominate in the smears and ghost cells are not easily identified. In such cases, careful re-evaluation of FNA smears in the right clinico-radiological context is warranted before giving a false positive diagnosis of malignancy.

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Funding: None; Conflict of Interest: None Stated.

How to cite this article: Kishore M, Madaan GB. Pilomatricoma of cheek mimicking malignancy: A diagnostic challenge on Fine needle aspiration cytology. Indian J Case Reports. 2019;5(5):487-489.

Doi: 10.32677/IJCR.2019.v05.i05.028