Pilomatricoma: A diagnostic challenge both for the clinician and cytopathologist

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Abstract
Pilomatricoma or Calcifying epithelioma of Malherbe is an uncommon benign tumour arising from the hair follicle. It comprises about 0.12% of all skin tumours, and is seen to occur in head and neck region and upper extremities in the first two decades of life. It is usually misdiagnosed clinically and preoperatively. We present a case report of a 55 year female with a firm, non tender, non pruritic nodule in trunk region measuring 2x2 cms since the past 1 month. FNAC was not contributory. However, histopathology confirmed the diagnosis of pilomatricoma. This case report highlights that pilomatricoma needs to be considered in the differential diagnosis of all solitary skin nodules of all sites and age groups.

Keywords: Pilomatricoma, Calcifying epithelioma, Trunk.

Introduction
Pilomatricoma also known as calcifying epithelioma of Malherbe is a rare benign tumour which is seen to arise from the matrix cells of the hair follicle and accounts for only 0.12% of all skin tumours.[1] They are commonly asymptomatic, seen to arise in head and neck region and upper trunk area and show a slow but progressive increase in size.

Preoperatively, it can present as a diagnostic challenge and surgical surprise due to its deceptive appearance clinically, cytologically and on imaging which leads to its misdiagnosis commonly in clinical practice.[2] As there are not many reports of pilomatricoma published in literature, our experience with the present case highlights the unusual clinical presentation, gross appearance of the tumour as well as the preoperative diagnostic pitfall.

Case Report
A 55 year female came to the Surgical OPD with a lump in the back measuring 2x2x1 cms for 1 month duration. The lump was firm to hard, non tender seen in the subcutaneous tissue which though, had a limited mobility was not fixed to the underlying tissue. The skin overlying the lump did not show any ulceration/punctum or erythema. However an area of hypopigmentation was noted. No imaging study was done. Fine needle aspiration cytology (FNAC) was attempted once but it showed only the presence of few giant cells against a mildly necrotic background hence a diagnostic possibility of foreign body granuloma or foreign body reaction was suggested. The tumour was excised and it measured 3x2x2 cms in size with multiple projections in deeper tissues. On microscopy, the tumour was composed of basaloid cells and larger ghost cells or shadow cells. The ghost cells had indistinct cell boundaries with abundant eosinophilic cytoplasm. Areas of calcification and few multinucleated giant cells were seen.

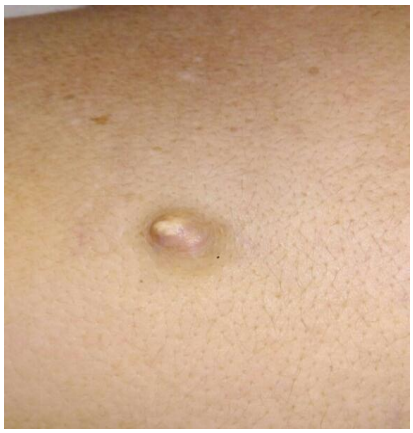

Fig. 1: Nodule over trunk measuring 2x2 cms

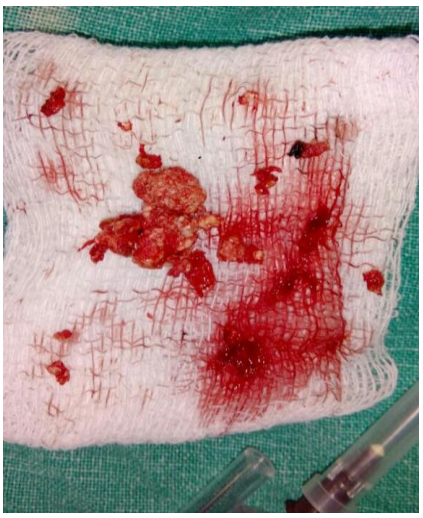

Fig. 2: Excised nodule measuring 3x2x1 cms

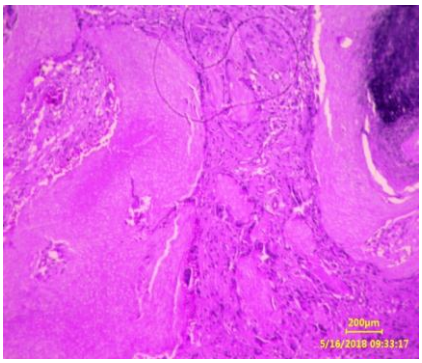

Fig. 3: Histopathology showing basaloid cells, ghost cells and focal calcification. H and E, 400X

Discussion

Pilomatricoma earlier known as Calcifying epithelioma of Malherbe was first described by Malherbe and Chenantais in 1880. The cell of origin was unclear but Forbes and Helvig in 1961 after an electron microscopic analysis of 228 tumours concluded it to be arising from the matrix cell which is the outer sheath of the hair follicle.[3] The common sites of occurrence are head and neck region and upper extremities. Though other sites have been reported, the location on the trunk region is uncommon with less than 15% of cases reported.[4]

The tumours may develop at any age, though a bimodal peak is seen during the first and sixth decades of life. However approximate 60% of cases are seen to occur in children and young adults within the first two decades of life.[5] The tumours are usually solitary, asymptomatic, well circumscribed, non tender subcutaneous tumours which are usually fixed to overlying skin. The skin may appear pale, teliangietactic, or blue black and may be thinned out with ulceration. The tumours are soft to firm depending on the degree of calcification present.

As pilomatricomas commonly occur in the head and neck region, they may be clinically misdiagnosed for various lesions like sebaceous cyst, epidermal cyst, dermoid cyst, chondroma, ossifying hematoma, degenerating fibroxanthoma, lipomas etc.[6]

Ultrasonography is usually not helpful in preoperative diagnosis as the appearances can be variable depending upon the degree of calcification.

Cytological diagnosis can be difficult as the diagnostic triad of basaloid cells, ghost cells (shadow cells) and foreign body giant cells may not always be present. Nucleated squamous cells or calcification if present are helpful pointers to the diagnosis. The presence of ghost cells or shadow cells is characteristic and a meticulous search is needed as these are difficult to be picked up on cytology.[1] FNAC in our case was misleading as the aspirate showed only multinucleate giant cells along with few squamous cells which were presumed to be contaminants. Ghost cells were not present in the aspirate in our case leading to an erroneous diagnosis.

On histopathology, the tumour usually appears as a localised lesion in the dermis containing islands of epithelial cells. The outermost cells are small with oval nuclei and basophilic cytoplasm and are thus called as basaloid cells. Towards the centre of the islands are ghost cells or shadow cells which are large, anucleate cells with abundant eosinophilic cytoplasm and large amounts of keratin. These areas represent maturation towards the hair cortex. Between these two layers of cells maybe interspersed a transitional layer which comprises of small nuclei. Calcification is usually seen. A surrounding inflammatory response comprising of inflammatory cells and foreign body giant cells is usually present around the tumour.

Pilomatricomas may be associated with other diseases like myotonic dystrophy, Gardner syndrome, Steinerts's disease, Turners syndrome and Sarcoidosis.[7]

Pilomatricoma needs to be differentiated from Pilomatrix carcinoma which is malignant. The presence of nuclear pleomorphism, atypical mitotic figures, central necrosis, lymphovascular invasion and infiltration into surrounding tissues favours the diagnosis of Pilomatrix carcinoma.[8]

Advances in molecular testing have made it possible to detect β catenin gene mutation in 75% of the pilomatricomas. This gene plays an important role in the development and diffrentaition of hair follicles. A strong β catenin expression was seen in the basaloid cells with the transitional cells and shadow cells being negative. Thus β catenin plays an important role in cell – cell adhesion and cellular proliferation and differentiation in the pathogenesis of pilomatricoma.[9]

The standard treatment of pilomatricoma is wide local surgical excision. Recurrences are reported to be less than 3%.[10]

Our experience with this case highlights the uncommon age and site of presentation. As it can occur at unusual sites where it is frequently misdiagnosed, it should be included as a differential diagnosis in all superficial lesions. A meticulous search for the characteristic cells would help in an accurate preoperative diagnosis of this uncommon entity.

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Conflict of Interest: Nil

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