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Case Report

Calcifying epithelioma of Malherbe of cheek: a diagnostic challenge

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ABSTRACT

Malherbe's calcifying epithelioma is a very rare benign tumour in area of cheek but generally present in first two decades of life. Fine Needle Aspiration Cytology (FNAC) and radiological imaging technique are not of great help in diagnosing this tumour. We hereby present a case of 15 year female with cheek swelling since 3 years which turned out to be pilomatricoma on histopathology of specimen of excisional biopsy.

Keywords: Malherbe's calcifying epithelioma, Pilomatrixoma, BCL-2, CTNNB1

INTRODUCTION

Malherbe's calcifying epithelioma is a very rare benign tumour which arises from the matrix cells at the base of hair. They generally present as benign swelling increasing in the size slowly and progressively. Pilomatrixoma, or calcifying epithelioma of Malherbe, was first described in 1880 by Malherbe and Chenantais.[1]

They are most commonly seen in head and neck region but the rarity of this tumour and inability for needle aspiration cytology or imaging test to detect this tumour pre-operatively presents as diagnostic challenge and surgical surprise.

CASE REPORT

15 year old female presented with a swelling in the left cheek since 3 years. The swelling was less than a centimeter to start with which progressively increased up to the size of about a cherry in 3 years. There was no obvious cosmetic deformity. The swelling could be examined only on palpation. On palpation, the swelling was hard and non-tender present in subcutaneous tissue. It was freely mobile without any fixity to underlying muscular tissue or overlying skin. Overlying skin did not show any punctum or erythema. There was no fluctuation or transillumination or pulsation. There was no intra-oral bulge or fistula.

On ultrasonography (USG), it showed a hypoechoic mass with posterior shadowing suggestive of hairy mass which has most probably calcified. Fine needle aspiration was attempted twice but showed only giant cells without providing diagnosis of the pathology.

Patient was taken for surgery in local anaesthesia with intra-venous sedation. An intra-oral approach was selected to prevent any scar on the face. The dissection was done and the mass was removed. Complete haemostasis was achieved and wound was closed by using 4-0 round body vicyrl in two layers.

On gross examination, the mass was pearly white in colour and hard in consistency. It was gritty from which various layers could be peeled off and was not calcified. On microscopic examination, it showed features of being pilomatrixoma.

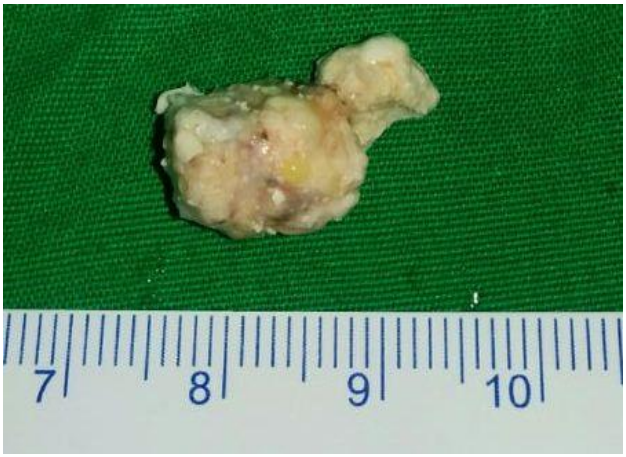

Figure 1: Gross specimen of Malherbe's calcifying epithelioma.

DISCUSSION

Pilomatrixoma is benign tumour arising from the matrix cells of hair. It was first described in 1880 by Malherbe and Chenantais.[1] In 1961, after histochemical and electron microscopic analysis of 228 such tumors, Forbis and Helwig found the cell of origin to be the outer root sheath cell of the hair follicle and proposed the name, pilomatrixoma, now called pilomatricoma.[2] These tumours depict bimodal peak presentation during the first and sixth decades of life, however, 40% of cases occur in patients younger than 10 years of age and 60% of cases occur within the first two decades of life.[3] There is a female pre-ponderance for the affection of the disease.[4,5] The most common site of affection is head and neck area with more than half the reported cases in this area. The most common site to be affected in the head and neck are frontal, temporal, peri-orbital and pre-auricular. In our patient, the site was cheek which is rare site for pilomatrixoma. The second most common region for affection is upper extremity.[6]

The basic pathology at the molecular level is an overactive proto-oncogene called BCL-2 suggesting that the normal process of cell death is suppressed and mutations in CTNNB1 suggesting loss of regulation of a protein complex called beta-catenin/LEF).[7] There is a school of thought that S100 proteins can be used as biochemical markers in characterization of pilomatrixomas.[8] Furthermore, investigators have shown that at least 75% of persons with pilomatrixomas who have examined have mutations in the gene CTNNB1; these data directly implicate beta-catenin/LEF misregulation as the major cause of hair matrix cell tumorigenesis in humans which is supported by the fact that the pilomatrixomas are strongly positive for LEF 1. Disorders that can be associated with pilomatrixoma include Gardner syndrome, myotonic dystrophy, Steinert disease, Turner syndrome, and sarcoidosis and Rubinstein-Taybi syndrome.

Major factors contributing to misdiagnosis include: cystic lesions with varying consistency, punctum like appearance (due to skin tethering), atypical location and absence of clinically recognizable calcification.[5]

The clinical differential diagnosis of head and neck pilomatrixoma includes sebaceous cyst, ossifying hematoma, brachial remnants, preauricular sinuses, adenopathy, giant cell tumor, chondroma, dermoid cyst, degenerating fibroxanthoma, foreign body reaction, and osteoma cutis.

Fine-needle aspiration cytology has been described as a preoperative diagnostic investigation. However, the results can be misleading if there are no ghost cells present in the aspirate.[10] USG is not very helpful in diagnosis but may show a hypoechoic lesion at the junction of the dermis and subcutaneous fat with focal thinning of the overlying dermis as was in our case. CT scan show a subcuraneous sweeling which does not enhance with contrast with variable amount of calcification. MRI will reveal uniform, homogeneously low to intermediate to high signal with variable contrast uptake on T1 weighted images and varies heterogenous with focal areas of hyperintensity on T2 weighted images. These findings do not help in diagnosing the pathology correctly on histopatholgy. The tumour is comprised of a basaloid proliferation which grows into structureless eosinophilic cells lacking nuclei called shadow cells or ghost cells. These areas represent maturation towards the hair cortex.

The standard treatment of pilomatrixoma is complete surgical excision. Wide excisions with margins of 1 to 2 cm are recommended for malignant variants to minimize the risk of local recurrence. The skin or the underlying tissue has to be excised if adherent to the swelling. Malignant transformation of the swelling is very rare but should be suspected in cases of local recurrences.

CONCLUSION

Pilomatrixoma are very rare benign tumours which may masquerade as other benign swelling that region. High clinical suspicion and excisional biopsy with immunohistochemistry typing are the true guides towards correct diagnosis. FNAC and radiological imaging are not of great value in suggesting the diagnosis. Surgical line of management is the gold standard for treatment of Malherbe's calcifying epithelioma.

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