

CASE REPORT

PILOMATRIX CARCINOMA IN A 4-YEAR-OLD CHILD WITH AN UNUSUAL PRESENTATION

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Pilomatrix carcinoma is a rare, locally aggressive variant of pilomatrixoma with a high rate of recurrence and risk of distant metastasis. We report an unusual presentation of a pilomatrix carcinoma in a 4-year-old male child who presented with recurrent lesions on his left cheek. At the age of 1 month of life, he presented with a soft tissue swelling on his left cheek. The lesion showed a circumscribed proliferation of basaloid cells with central areas of eosinophilic ghost shadow cells and intermediate cells. Basaloid nests showed round to oval, hyperchromatic nuclei with open nuclear chromatin, prominent nucleoli and frequent mitoses but no marked nuclear pleomorphism or infiltration was identified. The lesion recurred twice at the same site. Both recurrences showed similar morphology as the primary tumour however there were extensive areas of stromal necrosis, infiltrating edges, frequent mitoses with atypical forms, and lymphovascular invasion. There was no marked nuclear pleomorphism. Morphological features favoured a diagnosis of pilomatrix carcinoma. The child is still on follow-up and no recurrence has been identified to date. Pilomatrix carcinoma is rarely reported in infants. Due to its rarity, aggressive histological features may be missed.

Keywords: Pilomatrixoma; Pilomatrix carcinoma; Malignant pilomatrixoma; Child

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INTRODUCTION

Pilomatrixoma, or calcifying epithelioma of Malherbe, is a benign cutaneous adnexal neoplasm that shows differentiation towards the hair matrix of the hair follicles. These benign neoplasms are found primarily in the head and neck region and present as a slow-growing, hard, nodular lesion, in the subcutaneous tissue adherent to the skin.¹ Although benign pilomatrixoma can occur at any age, they have shown a bimodal age of presentation in the first and sixth decade of life, with the majority being reported before the second decade of life.²

Pilomatrix carcinoma is a rare variant of pilomatrixoma which was first described by Lopansri and Mihm in 1980 as a locally aggressive tumour. In the past, it was considered a low-grade malignant tumour with a high risk of recurrence following simple excision while the risk of metastasis was quite low. However distant metastasis is now well documented with multiple reported cases of lymph node metastasis and systemic metastasis, mainly to lungs.³

Compared to pilomatrixoma, pilomatrix carcinoma also shows a bimodal age of presentation but their peak age of presentation is in the 5th to 7th decade of life.⁴ They show infiltrative nests of basaloid cells with marked nuclear pleomorphism and abundant or brisk mitotic activity (8-62/ high power field). These neoplasms often show central areas of necrosis and vascular and lymphatic

invasion. We herein report a case of a recurrent tumour at cheek with follicular differentiation in a 4-year-old child that started at 1-month of age.

CASE REPORT

A 4-year-old child presented with recurrent lesions in his left cheek. He was a known case of acute lymphoblastic leukaemia/lymphoma (B-ALL) and was undergoing treatment. We received total 12 blocks of his lesions at our hospital for review.

The child presented at the age of 1 year in 2018 with a painless, palpable soft swelling on his left cheek. According to the parents, the swelling was present for one month of age. The lesion measured 1.8×1.5×1.3 cm. The lesion was excised and reported at an outside institute as pilomatrixoma. We received 2 blocks of this lesion. H&E stained slides of this lesion showed an unencapsulated neoplasm showing lobulated proliferation of immature basaloid cells in sheets and nests. The cells had moderate cytoplasm. Nuclei were round and oval with prominent nucleoli. Mitotic figures were easily appreciated along with areas of necrosis. Very focally necrosis was associated with necrotic debris and it appeared different from the necrosis associated with ghost cells. Adjacent to these basaloid cells, clusters of squamoid cells were seen with trichilemmal type of keratinization. Multinucleated giant cell reaction around the keratinous material and focal areas of dystrophic calcification was seen. No lymphovascular invasion was identified. The tumour extended to the margin of resection (Figure-1).

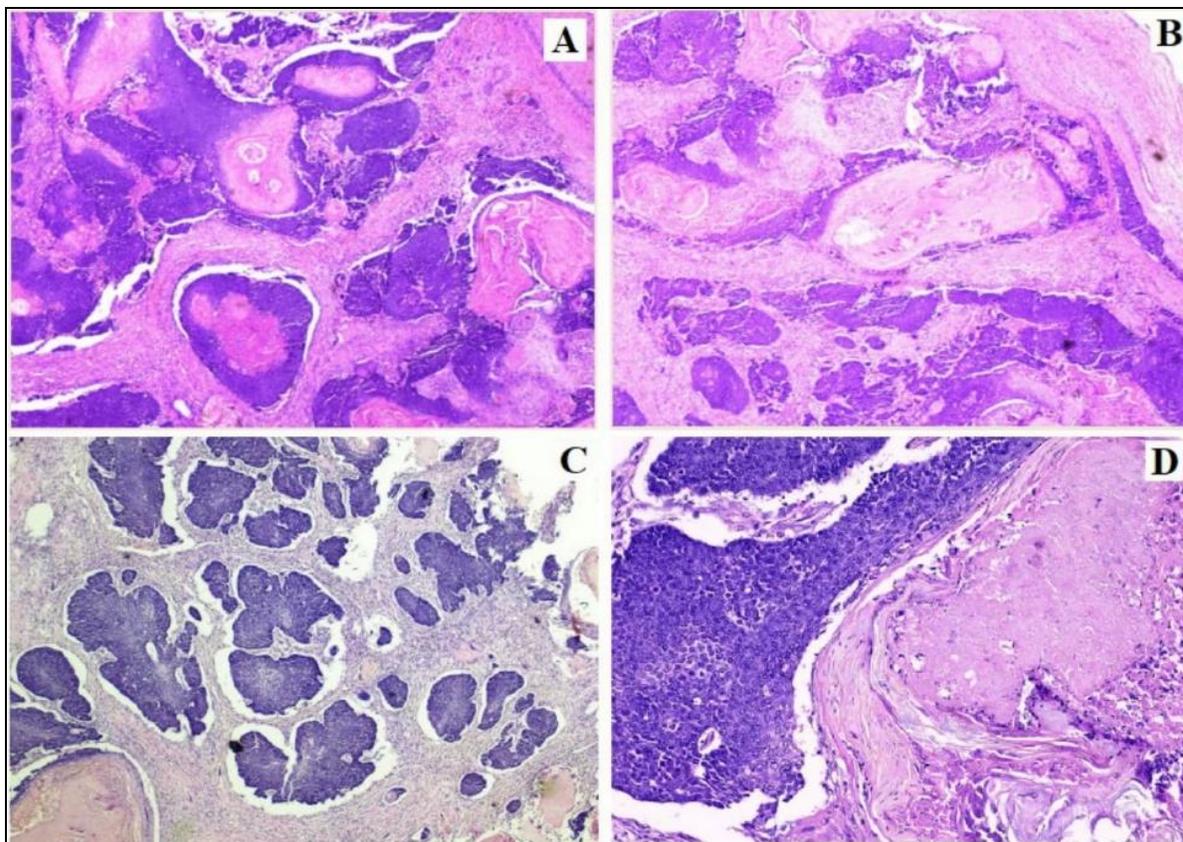


Figure-1: (A) Primary lesion exhibiting nests of basaloid cells with central trichilemmal type of keratinization (H7Ex 20X); (B) Primary lesion exhibiting circumscribed proliferation of basaloid (H7E x 10X); (C) First lesion exhibiting proliferation basaloid cells with few foci of shadow cell (H&E x 10X); (D) Basaloid cells with areas of necrosis and shadow cell (H&E x 20X)

The child presented again in September 2019 with recurrence at the same site. The lesion measured 2.5×2.0×2.0 cm. Excisional biopsy of the recurrent lesion was done and reported at some other institute as proliferating pilomatrixoma. We received 8 paraffin-embedded blocks of this lesion. H&E stained slides showed a neoplastic lesion focally covered by skin. Extensive ulceration with acute and chronic inflammatory infiltrate and granulation tissue formation was noted. The underlying dermis showed a large lobulated tumor identical to the initial tumor. The neoplasm showed solid nests of basaloid cells intermixed with hyalinized ghost cells. There was, however more pronounced necrosis compared to the initial tumor. Basaloid areas showed more solid proliferation of immature cells exhibiting round nuclei with prominent nucleoli and moderate cytoplasm. Prominent mitotic figures were seen; however, no atypical mitoses were identified. Central necrosis was also identified in the hyalinized keratinous material along with extensive areas of stromal necrosis. Focal areas of dystrophic

calcification were seen (Figure-1). No lymphovascular invasion was identified.

In February 2020 the child developed a second recurrence at the same site. The tumor was excised with wide clear margins. It was reported from an outside institute as pilomatrical carcinoma. We received 2 blocks of this lesion which showed a tumour similar to the ones seen previously. However, the nests and strands were smaller and there was a higher proportion of immature hyperchromatic cells. Mitotic activity was brisk with occasional atypical forms.

There were areas of squamous differentiation with abrupt keratinization. Background stroma appeared sclerotic and scarred, which is a reflection of recurrence. The neoplasm showed infiltration into the adjacent adipose tissue. Foci of lymphovascular invasion were identified which were fairly prominent and highlighted on immunohistochemical stain CD31 (Figure-2). Morphological features were consistent with pilomatrix carcinoma. The patient is on a follow-up to date and no recurrence has occurred since. CT scan of head and neck and chest was negative for metastasis.

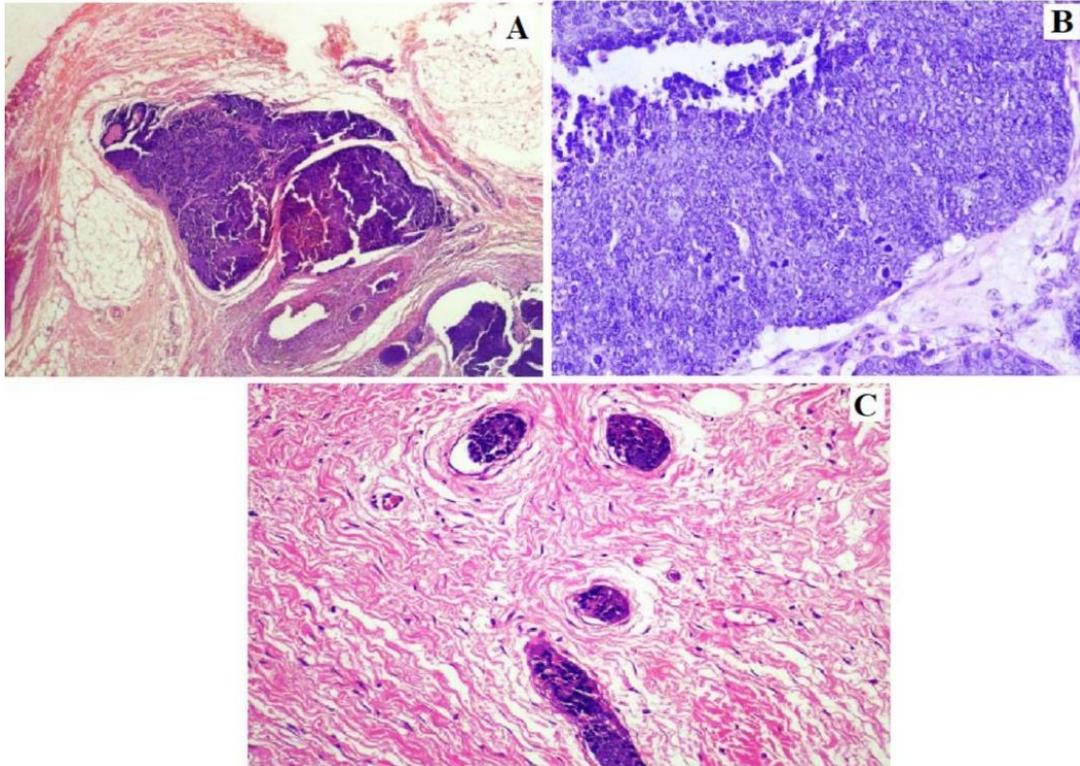


Figure-2: (A) Second recurrent lesion exhibiting infiltration into the adjacent sub-cutaneous tissue (H&E x 10X); (B) Second recurrent lesion showing mild nuclear pleomorphism and numerous mitotic figures (H&E x 40X); (C) Second recurrent lesion showing vascular invasion (h&E x 40X)

DISCUSSION

The current case posed a diagnostic challenge owing to the rarity of pilomatrix carcinoma in children, absence of marked nuclear pleomorphism, and unfamiliarity with the intermediate pilomatrical categories, i.e., proliferating and aggressive pilomatrixoma. Although the morphology in the initial lesion and recurrences was the same showing predominance of basaloid nests, numerous mitoses, infiltrative borders and necrosis, they became more prominent in the last recurrence.

Pilomatrical neoplasms are the most common cutaneous neoplasms of childhood. While most of these neoplasms are benign pilomatrixomas, rarely, pilomatrix carcinomas are also reported.³ In 12 years literature review of pilomatrix carcinoma done by Jone *et al*, the minimum age of presentation was 8 years.⁴ There is only one case report by Kim *et al* who reported pilomatrix carcinoma in an 8 months old infant who presented with a 3-month history of an erythematous nodule in his right cheek.⁵

In 1997 Kaddu *et al*. retrospectively evaluated pilomatrixoma and found unusual histological features not typical for a benign pilomatrixoma. They referred to these cases as proliferating pilomatrixoma. These uncommon forms

of pilomatrixoma showed features intermediate between malignant pilomatrix carcinoma and benign pilomatrixoma.⁶ They showed relative symmetry, a sharp circumscription of the lesion, basaloid aggregates with smooth contours and lack of perineural and vascular invasion. However, the presence of variable atypia of the basaloid cells, increase mitoses (1 to 6 per HPF) and areas of necrosis confuse them with pilomatrix carcinoma.

Since then, multiple case reports of this entity are published. However, due to its rarity differentiating histologic features from pilomatrix carcinoma are not well defined and lacks standardization.⁷ Most of the reported cases of proliferating pilomatrixoma exhibited a focal infiltrative pattern, foci of comedonecrosis, variable nuclear atypia and increased mitotic activity. None of these cases showed vascular or lymphatic invasion.⁷ It is important to recognize this variant due to its potential for local recurrence compared to conventional or benign pilomatrixoma. Moreover these intermediate neoplasms have low metastatic potential, therefore differentiation from other malignant tumours such as basal cell carcinoma and pilomatrix carcinoma is important.⁸

Compared to benign pilomatrixoma and proliferating pilomatrixoma, pilomatrix carcinoma

exhibits anaplastic, pleomorphic nuclei with prominent nucleoli.⁴ Papadakis et al observed certain histological features to differentiate between proliferating pilomatrixoma and pilomatrix carcinoma. Proliferating pilomatrixoma shows slightly atypical nuclear features with an expansile growth pattern while necrosis and vascular invasion is rarely observed. In contrast, pilomatrix carcinoma demonstrates marked nuclear pleomorphism and anaplasia, increased mitotic activity and necrosis.^{3,9}

Surgical excision with wide clear margins has shown a low rate of recurrence. Hardisson et al have reported that tumours that were excised with a margin of 5–10 mm did not show any recurrence, therefore they recommended a minimum of 5 mm as an adequate margin for pilomatrix carcinoma.¹⁰

CONCLUSION

Characterizing the current lesion into a proliferating pilomatrixoma and malignant pilomatrix carcinoma was a problem owing to the absence of well-defined histopathological criteria and nomenclature. Although there was no marked nuclear pleomorphism as reported in the literature, the presence of infiltrating margins, increased mitotic activity, foci of necrosis and lymphovascular invasion favored a diagnosis of malignant pilomatrix carcinoma which is a very rare presentation in a child.

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