

## Pilomatrix carcinoma - pilomatrixoma: Two ends of the disease spectrum

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### Abstract

Pilomatrixoma, pilomatrix carcinoma, are neoplasms of skin appendage with differentiation towards hair follicle matrix cells. They represent two ends of the spectrum with varying clinical and pathological characteristics. Pilomatrix carcinoma is a rare malignant neoplasm, having a tendency to recur locally, with a rare propensity for distant metastasis.

There is a need to differentiate the two entity, for appropriate selection of treatment. We describe a case of a pilomatrixoma in a 62 year old gentleman over the right chest along with a review of pathology and treatment options.

**Keywords:** Pilomatrix Carcinoma, Pilomatrixoma, Cutaneous neoplasms.

**Key Message:** Pilomatrix carcinoma is a rare cutaneous malignancy affecting with a predilection to elderly, male population. It can be differentiated from benign pilomatrixoma by histopathology. Surgical excision with wide margin is the current standard of care, associated with least recurrence. Adjuvant radiation may be offered for margins positive resection or in inoperable cases.

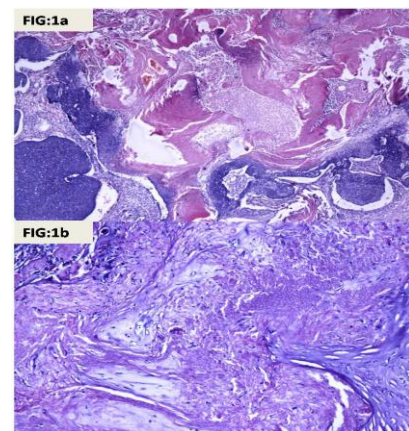
### Introduction

Pilomatrixoma, an uncommon neoplasm of the skin hair follicle, arises from the cortical cells of root of hair follicles. Pilomatrix carcinoma, the malignant counterpart, is a very rare entity with only around 100 cases described in English literature till date.<sup>1</sup> Clinical differentiation of the two is difficult, and pathological distinction based histomorphological features is the key to diagnosis.<sup>2</sup> Surgical resection is the primary treatment of choice, though radiation has been tried in a few cases with limited success.

### Case History

A 63 year old gentleman presented with gradually enlarging swelling over right side of chest in infraclavicular region over 5 months. Clinical examination revealed a globular 5x5 cm, firm, well defined, bosselated swelling in the right anterior chest. Skin over the swelling was intact however lump was tethered to pectoralis major muscle. No axillary or neck nodes were palpable. Magnetic resonance imaging (MRI) of chest revealed a soft tissue mass in the subcutaneous plane, abutting onto the underlying pectoralis major muscle measuring 31.8x31.4x22.5 mm, with No significant axillary or neck nodes. Positron emission tomography (PET) scan revealed minimal uptake in the mass [SUV 2.3] with no other sites of uptake. Fine Needle Aspiration Cytology (FNAC) from the mass was suggestive of non contributory. Patient underwent wide local excision with margins along with a cuff of pectoralis major

muscle. Frozen section from margins was negative for malignancy. Final histopathology revealed a 2.8x.3.0x2.1 cm tumour with clearance all around of 1.5 to 2.5 cm. Light microscopy revealed tissue lined by stratified squamous epithelium, subepithelium showed a tumor composed of nest of small basaloid cells showing hyperchromatic nuclei with abrupt keratinization, foreign body type of giant cell reaction, no mitosis and focal calcification, suggestive of pilomatrixoma (Fig. 1). At 5 years after surgery patient is on follow up, asymptomatic without recurrence.



**Fig. a): Hematoxylin and eosin staining : 4X zoom, b) 10 X magnification**

**Legend a): The figure shows H basaloid cells with transition to a large mass of ghost cells.**

**Fig. b): Ghost cells seen at 10 X magnification.**

### Discussion

Pilomatrixoma, benign neoplasm of hair follicle cells, occurring in young adults, with nearly 45 % cases occurring before the age of 18.<sup>3,4</sup> It is more common among women, in head and neck region.<sup>5</sup> It usually occurs as solitary superficial firm skin lesion, however multiple lesions have been found in association with

familial syndromes like myotonic dystrophy, gardners syndrome

Pilomatrix carcinoma, occurs more commonly among the elderly, predominantly men], affecting face, neck, scalp, back, shoulder/arms, leg in decreasing order.<sup>1</sup> It can arise de novo or can occur from transformation of pilomatrixoma or following pilomatrixoma resection.<sup>6</sup>

Distinction between pilomatrixoma and pilomatrix carcinoma is predominantly based on histomorphology. Pilomatrixoma is composed of two groups of cells - basaloid and anucleated 'shadow' or 'ghost' cells. Histomorphologically it is characterized by the presence of uniform basaloid cells, with prominent nucleoli showing squamous differentiation and degeneration, centrally within the tumor, leaving anucleated ghost [shadow] cells,<sup>7,8</sup> pilomatrix carcinoma is characterized by the presence of infiltrative growth pattern, atypical mitotic figures, tumor necrosis, nuclear pleomorphism, dense desmoplastic stroma, prominent lymphoplasmacytic infiltrate and perineural or perivascular invasion.<sup>2</sup> Immunohistochemical or molecular markers are not useful in differentiating benign and malignant variants.<sup>4,5</sup> At the molecular level, mutation is observed in CTNNB1 gene encoding Beta Catenin - Wnt cell regulation pathway, observed in both pilomatrixoma and pilomatrix carcinoma, implying a common initial pathogenesis and a possible explanation for occurrence of pilomatrix carcinoma arising from pilomatrixoma.<sup>1</sup>

On fine needle aspiration cytology, the presence of malignant population of basaloid cells, with presence of ghost cells and keratin clumps indicates the possibility of pilomatrix carcinoma pathologically two other variants, aggressive pilomatrixoma and proliferating pilomatrixoma are also described in literature.

Clinically pilomatrix carcinoma has a high tendency to recur locally, higher recurrence rates with simple excision than with wide local excision (64% vs 17%).<sup>9</sup> Distant metastasis can occur via blood and lymphatic spread with almost 10% cases reported showing metastatic disease, with lungs being common site of metastasis.<sup>9</sup>

Surgical resection has been the accepted standard treatment for pilomatrix carcinoma. Because of the rarity, lack of large series, optimal margin of resection is unclear and a margin of 5–10 mm all around has been recommended Mohs micrographic surgery has been to be successful in tumors of face and back, without any evidence of recurrence.<sup>10</sup>

The site of tumor, latency period, size of tumor had no association with recurrence, while higher degree of anaplasia, increased depth of tissue infiltration had been associated with increased incidence of recurrence.<sup>9</sup> Thus it is difficult to predict the prognosis of pilomatrix carcinoma, except for the fact that wide local excision provides the least chances of recurrence.

Radiation can be used as postoperative adjuvant therapy, as definitive treatment especially in unresectable tumors. In recurrent tumors, good local control has been achieved in literature with radiation [external beam/ brachytherapy], with no effect on systemic progression.

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