Skin: Pilomatricoma

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Identity

Alias
Pilomatrixoma; Trichomatricoma; Calcifying Epithelioma of Malherbe

Note
Benign skin tumor with follicular differentiation arising from hair cortex cells.

Clinics and pathology

Disease

History:
- 1880: Malherbe and Chenantias described a benign neoplasm of the skin which they thought to be arising from sebaceous glands and called it as "l'epithelio me calcifie des glandes sebacees". Since then, it has been termed as Calcifying Epithelioma of Malherbe.
- 1922: Dubreuilh and Cazenave gave the unique histopathologic description of this neoplasm consisting of basaloid and shadow/ghost cells.
- 1942: Turhan and Krainer determined that the origin of this neoplasm to be from hair matrix cells.
- 1961: Forbes and Helwig renamed the neoplasm as "Pilomatrixoma" to aptly describe its origin from hair follicle matrix cells and avoiding the word epithelioma which carried the connotation of malignancy.
- 1990: Headington has used the name "trichomatricoma" to describe these neoplasms.

Embryonic origin
Pilomatricoma is thought to arise from hair matrix cells.

Epidemiology
Pilomatricoma accounts to 20% of all hair follicle related tumors in most series. Therefore, it can be considered as commonest hair follicle tumor. It predominantly affects the children and young adults with 40% of cases occurring before the age of 10 years and 60% of cases before the age of 20 years. However, it has also been reported in older adults. A bimodal age distribution of first and sixth decades has been reported by several authors. They exhibit a definite female preponderance with an M: F ratio of around 2:3 and are commonly encountered in whites. The most frequent anatomical location is in the head and neck region (>50%) followed by upper extremity, trunk and lower extremity.

Clinics
Pilomatricoma usually presents as a solitary, slow growing, asymptomatic, superficial/deeply seated, freely mobile, firm to rock hard mass ranging in size upto 0.5-5cm in diameter. The overlying skin can be normal or exhibit a bluish-red discoloration or even ulceration rarely. The diagnosis is usually suspected based on palpation of superficial rock hard mass and confirmed by histopathologic examination.
The tumor if stretched may show several facets and irregular angles, referred to as 'tent sign'.

Multiple tumors and familial cases also occur. They are usually associated with Gardner's syndrome, Myotonic dystrophy, Steinert's disease, Rubinstein-Taybi syndrome, Turner's syndrome and sarcoidosis. Occasional variations like aggressive, symmetrically localized and giant lesions have also been reported.

Radiology is of little diagnostic value. It is mainly used to differentiate pre-auricular tumors from parotid tumors and for evaluating large and aggressive tumors. Plain X rays reveal nonspecific calcification while ultrasound shows a well defined round hyperechogenic mass with post dense acoustic shadow. CT scan and MRI reveal a sharply demarcated, subcutaneous opaque lesion that does not enhance on injection of contrast material or small areas of signal dropout consistent with presence of calcifications.

**Cytology**

The aspirates usually reveal two types of cells i.e. anucleated shadow/ghost squamous cells and basaloid/basophilic squamous cells and nucleated squamous cells and areas of calcification. Generally, as the neoplasm ages, the aspirate will consist predominantly of shadow cells with few cells of other categories.

**Anucleated shadow cells**: These are highly characteristic of pilomatricoma. They are present either as isolated cells or in small clusters. They are generally small, possess distinct cell borders with pink keratinized cytoplasm. A clear usually unstained area occupies the site of the nucleus but occasionally, a faint trace of the nuclei can be seen.

**Basaloid cells**: These cells are arranged in tight clusters or in sheets. They were oval to polygonal with poorly defined cell borders and scanty cytoplasm, high nuclear-cytoplasmic ratio containing vesicular to hyperchromatic nuclei and occasional nucleoli.

**Additional findings**: Chronic inflammatory cells, multinucleated giant cells, large fragments of calcific deposits.

**Pathology**

The neoplasm is usually well encapsulated containing irregularly shaped, lobulated islands of cells separated by fine, fibrovascular connective tissue stroma. The islands demonstrated two distinct cell populations i.e. comprising of basaloid cells located towards the periphery and the ghost/shadow cells occupying the central portion.

**The basaloid cells** are darkly stained, round or elongated, with indistinct cell borders and minimal cytoplasm. They contain round to oval nuclei with high cellular content. Anucleated cells and cellular debris are also present.

**The ghost/shadow cells** in the center of the islands have abundant, pale, eosinophilic cytoplasm with well defined cell borders and central clear area. Most of them show only faint traces of nuclei. A transition from basaloid to ghost cells is seen in most areas which may be abrupt or gradual.

The shadow cells are formed due to keratinization of basaloid cells and tend to increase in number as the neoplasm ages. Secondary histopathologic features include hemorrhage, foci of calcification either in the form of fine basophilic granule or a large fragment of amorphous basophilic material, ossification, foreign body giant cells with inflammatory cells, whorls of keratin, melanin pigmentation, myxoid change, necrosis etc.

**Treatment**

Surgical excision is the treatment of choice.
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A: Photomicrograph depicting the encapsulated pilomatricoma composed of islands of cells with peripheral, dark basaloid cells; and pale central areas of shadow cells, along with areas of keratinisation. Inset: Magnified view of basaloid and ghost cells.

B: Photomicrograph depicting the encapsulated pilomatricoma composed of islands of cells with peripheral, dark basaloid cells; and pale central areas of shadow cells, along with areas of keratinisation. Areas of cystic degeneration are also evident.

C: Photomicrograph demonstrating peripheral darkly stained basaloid cells and central pale shadow cells.

D: Showing areas of ghost cells, keratin material, giant cell reaction and inflammatory infiltrate.

E: Showing whorls of keratin in the center of the islands.

Evolution

Most pilomatricomas increase slowly in size, some cease to grow to a small size while others grow up to a size of 15 cms. They can rupture spontaneously and discharge a chalky material. Hemorrhage within a pilomatricoma can lead to rapid enlargement. Malignant transformation into pilomatrical carcinoma has also been rarely described and has to be suspected if there is evidence of recent rapid growth, atypically large lesion, and ulceration, fixity to adjoining structures, infiltration or high mitotic activity.

Prognosis

They seldom recur, if the excision is complete. Local recurrence may develop due to incomplete excision. If multiple recurrences are noted, excision with margins has to be done to rule out the possibility of pilomatrical carcinoma.

Genetics

Note

Most cases of pilomatricoma occur sporadically without an apparent genetic predisposition.
However, few are associated with the following known genetic syndromes:
- Gardner’s syndrome
- Turner’s syndrome
- Myotonic dystrophy
- Steinert’s disease
- Rubenstein Taybi syndrome.

**Genes involved and proteins**

**CTNNB1**

**Location**
3p21

**Note**

Pilomatricomas usually contain mutation of CTNNB1, a gene encoding 92 kDa protein beta-catenin which is a multifunctional protein related to the adherens junction, signal transduction and is a key molecule of cell proliferation. It is central to epithelial architecture and regulates the polarity of cells and tissues. Beta-catenin stabilization may play a key role in epidermal signaling leading to hair development, and its aberrant activation may be implicated in formation of hair tumors. Beta-catenin gene mutations of the hair matrix cells stabilize beta-catenin proteins which accumulate in the cytoplasm and translocate to the nucleus. These beta-catenin proteins in the nucleus in turn activate gene transcription via Wnt-TCF-Lef-1, resulting in abnormal matrical cell proliferation and formation of pilomatricoma.

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This article should be referenced as such: