Pilomatricoma.

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Pilomatricoma, also referred to as pilomatrixoma and calcifying epithelioma of Malherbe, is a benign dermal-subcutaneous tumor derived from the matrix of the hair follicle. Its development is associated with a known mutation in the CTNNB1 gene, the gene that encodes for beta-catenin. Pilomatricomas are relatively common tumors. They usually arise during the first 2 decades of life, and they have no predilection for either sex.

The most commonly affected sites are the head and neck and the upper limbs. The lesion presents as a solitary, rubbery to hard mass that often leads to a “tent sign” appearance to the skin. The occurrence of multiple tumors is rare; when they do occur, they are usually syndrome-associated lesions. Simple excision is curative, although some rare cases undergo malignant transformation to pilomatrical carcinoma.

These tumors can be as large as 3 cm in diameter. Their cut surface is gritty to chalky as a result of tumor calcification. Tumors are well-circumscribed, dermal to subcutaneous nodules composed of several components, including basaloid proliferation, shadow cells, dystrophic calcifications, and foreign-body giant-cell reactions (figure 1).

Figure 1. A: Low-power photograph shows a basaloid tumor deep in the dermis and subcutaneous tissues. B: A basaloid proliferation juxtaposed to a ghost cell fragment exhibits early signs of calcification. Numerous giant cells are present between the fragments.

The basaloid cells are tightly cohesive, usually more prominent at the periphery of the tumor, and composed of many layers of small, monotonous cells. The cells are arranged in a syncytium with indistinct cell borders and a very high nucleus-to-cytoplasm ratio (figure 2). They have a high mitotic index, and they merge imperceptibly to abruptly with the keratinizing shadow cells.
Figure 2. A: High-power view shows the basaloid proliferation gradually blending into the keratinizing eosinophilic shadow-cell area. The nuclei are pyknotic just as they transition to the shadow cells (upper). B: A syncytium of basaloid cells exhibits a very high nucleus-to-cytoplasm ratio and numerous mitoses.

The shadow cells have abundant eosinophilic cytoplasm and a negative space where the nucleus was once located (figure 2). As lesions age, the proportion of shadow cells increases. The dystrophic calcifications begin within the shadow cells as fine granules (figure 1). They may ultimately become the dominant finding, resulting in ossification in up to 20% of cases.

Tumors are frequently cystic and might have ruptured, demonstrating a well-developed, foreign-body, giant-cell reaction. The shadow cells are usually part of the material that is destroyed by the giant-cell reaction.

In rare cases, melanin and extramedullary hematopoiesis may be seen. There is a prominent nuclear reaction with beta-catenin, but this is usually unnecessary for the diagnosis. The diagnosis is usually straightforward, although the basaloid population on fine-needle aspiration samples may be misinterpreted as a carcinoma.

Histologically, the differential diagnosis includes basal cell carcinoma (lacks shadow cells; shows clefting around basaloid groups), neuroendocrine carcinoma such as Merkel cell carcinoma (lacks shadow cells; shows CK20 immunoreactivity), and a proliferating trichilemmal cyst (usually large squamous cells and no shadow cells).

Suggested reading

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