

Pilomatrixoma: A Benign Appendageal Tumor not Uncommon in Children

Pilomatrixoma is a benign appendageal tumor, accounting for 1% of all benign skin lesions (1). It is mostly seen in children and young adults up to 20 years of age, the second peak being in the sixth and seventh decade of life. Pilomatrixoma usually manifests as a solitary, asymptomatic nodule on the head and less frequently on the upper and lower extremities. The diagnosis is commonly based on biopsy and histological findings of basaloid cells with calcium deposits within a fibrin capsule. The therapy of choice is complete surgical excision with a wide peripheral edge, which has a low probability of recurrence.

A 10-month-old male infant was referred to pediatric oncologist due to the formation located in the left temporal region, observed by his parents a month ago and associated with insect bite. The mass was painless, occasionally reddish-bluish, and had not changed in size. On examination a superficial, well circumscribed, and moderately firm lesion 1.5 cm in diameter was seen and palpated in the left temporal region (Figure 1). Plain radiography revealed no bone lesions. Ultrasonography demonstrated an ovoid, well-defined hypoechoic formation 1 cm in diameter, suggesting granuloma. Fine needle aspiration (FNA) was performed. Cytomorphologic analysis revealed the presence of abundant clusters of small



Figure 1. A ten-month-old boy with an asymptomatic solitary nodule in the left temporal region.

cells with denting nuclei and basophilic cytoplasmic protrusions surrounded by extracellular pink fibrillary substance, some phagocytes, lymphocytes, granulocytes, scantily calcificated detritus, and scarce squamous cells, characteristic for pilomatrixoma. A complete surgical resection was done. Histopathological findings confirmed the diagnosis (Figure 2). Eight months after the excision there were no signs of local recurrence.

Calcifying epithelioma was first described by Malherbe and co-workers in the 19th century (2). The tumor was later called pilomatrixoma to reflect the more accurate description of the origin from the hair follicle germinal matrix center. Pilomatrixoma is a benign solitary tumor usually occurring on the head. Multiple presentations have been described, sometimes in more family members, mainly in association with myotonic dystrophy (3). Pilomatrixoma typically presents as a firm, hard mass, with reddish purple discoloration of the skin resulting from intrinsic hemorrhage. It usually manifests as an asymptomatic nodule and rarely causes pain or itching due to ulceration and inflammation. The growth is rather slow, up to a

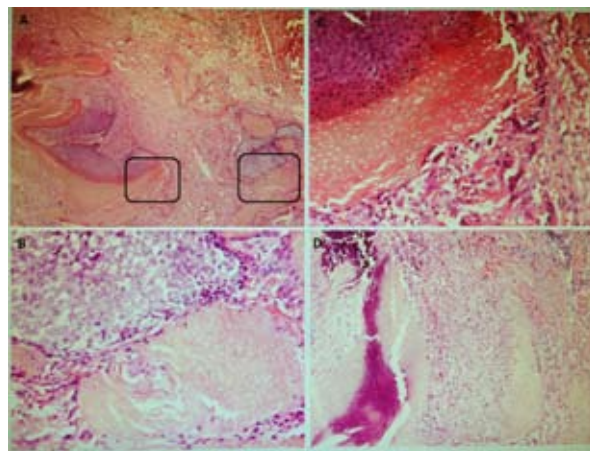


Figure 2. Pilomatrixoma: (a) lower power view shows typical biphasic population; (b) with maturation the cells become larger with abundant eosinophilic cytoplasm and nuclear piknosis; (c) foreign body reaction and (d) calcification is commonly present.

few centimeters; rarely, giant pilomatrixomas have been reported.

The majority of cases are diagnosed in the first two years of life. The sex distribution is still unclear, but slightly greater preponderance in males has been reported (4).

Although genetic predisposition most likely plays a role in the pathophysiology of tumors, the etiology has yet to be understood. The mutations in the CNN-BT1 gene and beta-catenin/LEF dysregulation have been identified as the major cause of tumorigenesis (5). In a few cases, a preceding insect bite or trauma have been reported.

Although clinical presentation itself suggests pilomatrixoma, and ultrasonography is a helpful diagnostic tool, misinterpretation of the lesion as an epidermal inclusion cyst, dermoid cyst, giant cell tumor, foreign body granuloma, and many others is frequent (6). FNA cytological features of shadow cells, primitive basaloid cells with a high nuclear/cytoplasmic ratio and foreign body giant cells are characteristic and allow a correct diagnosis (7). Nevertheless, misdiagnosis is frequent. Definite diagnosis is established by biopsy and histopathological findings of ghost cells, calcium deposits, and foreign body giant cells (7).

Spontaneous regression has not been observed. Complete surgical excision is the treatment of choice. Local recurrence seldom appears and is most likely due to incomplete resection. Malignant alteration has rarely been documented. It has been described in older men whose histopathological features included cellular pleomorphism, frequent mitotic figures and atypias (1).

Our patient had a slow-growing asymptomatic head lesion. An insect bite was clearly noted in the medical history. FNA proved a reliable method for preoperative diagnosis of pilomatrixoma.

We suggest that pilomatrixoma should be considered in the differential diagnosis of solitary adnexal tumors in children. Although rare, local recurrence can occur and patients should be monitored.

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Received: January 19, 2015

Accepted: October 20, 2015