

## Case Report

**J Chin Med Assoc**

2004;67:633-636

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### Key Words

calcifying epithelioma;  
myotonic dystrophy;  
pilomatrixoma

## Pilomatrixoma of the Head and Neck

Pilomatrixoma is a benign skin appendage tumor that commonly occurs as a solitary lesion and is not usually hereditary. There is evidence to suggest that patients with a family history of multiple pilomatrixomas have a high probability of autosomal dominant disorders such as myotonic dystrophy, Gardner syndrome, and Rubinstein-Taybi syndrome. In January 2004, a case was reported of an 8-year-old girl with 2 progressively enlarged facial masses. One of them was excised and diagnosed as pilomatrixoma. Coincidentally, her 13-year-old sister had the same type of tumor when she was 4 years old. We report this unusual case and review the literature. Pilomatrixoma has not been widely reported in the head and neck surgery literature. This benign tumor may be misdiagnosed as a carcinoma, resulting in unnecessary aggressive therapy. Otolaryngologists should therefore note the clinical and pathologic characteristics of these symptoms.

**P**ilomatrixoma (calcifying epithelioma of Malherbe), was first described by Malherbe and Chenantais<sup>1</sup> in 1880 as a calcified tumor, originating from the sebaceous glands. Later, Forbis and Helwig<sup>2</sup> further developed the histochemical and electron microscopic studies of pilomatrixoma and discovered that the cell of origin is the outer sheath cell of the hair follicle root. One series found a pilomatrixoma for every 824 dermatohistopathologic specimens. Multiple lesions are found in 3.5% of the reported cases.<sup>3</sup> Familial pilomatrixomas and multiple familial pilomatrixomas are even rarer.<sup>4</sup> Patients usually experience single pilomatrixoma, and when multiple, they may be associated with other diseases or syndromes.

Pilomatrixomas occur most often in the first 2 decades of life.<sup>5</sup> There appears to be a 3:2 female:male incidence ratio, with Caucasians being the most affected group.<sup>3</sup> The most common location of tumor is in the head and neck region. The prognosis is typically good, and the treatment of choice is surgical removal.

Although pilomatrixomas are easily recognized by dermatologists and pathologists, otolaryngologists are usually less familiar with them.

### CASE REPORT

In January 2004, our otolaryngology department examined an 8-year-old girl with 2 facial masses that had been growing for 6 months. Physical examination revealed 2 firm, painless nodules measuring, respectively, 1.5 × 1.5 cm and 1.0 × 0.5 cm in size. The nodules moved freely over the underlying subcutaneous layer on the left eyebrow tail (Fig. 1) and preauricular (Fig. 2) area, respectively. The overlying skin was elevated, flesh-colored and soft. The remainder of the examination was unremarkable.

The patient had a history of a painless subcutaneous nodule on her neck when she was 4 years old. It had been excised and diagnosed as pilomatrixoma.

The patient was otherwise healthy. A similar nodule was discovered in her 13-year-old sister, and was treated with surgical excision. The histopathological examinations confirmed the diagnosis of pilomatrixoma. No family consanguinity was recorded.

The tumor on the 4-year-old patient's eyebrow tail was excised under general anesthesia. After reviewing the patient's history and clinical findings, we decided not

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Received: May 28, 2004.

Accepted: August 23, 2004.

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to remove the benign lesion on the preauricular area to avoid permanent scarring.

Macroscopically, the tumor was 1.2 × 1.0 cm in size (Fig. 3). In cut sections, it was gray-white in color. Microscopic examination revealed histopathological features of pilomatrixoma, which was composed of fibrous stroma and surrounded by irregular islands of basophilic cells and shadow cells (Fig. 4).

Venous blood samples for DNA analysis were sent to



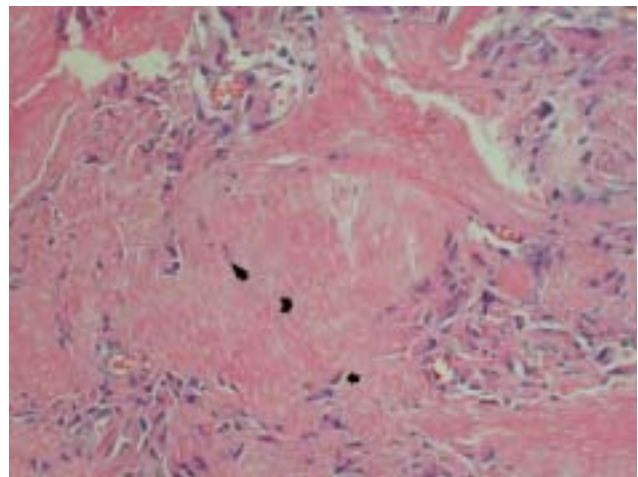
**Fig. 1.** Clinical photography demonstrating the appearance of pilomatrixoma occurring in the left eyebrow tail (arrow).



**Fig. 2.** Solid mass on the left preauricular region (arrow).



**Fig. 3.** Macroscopic appearance of the tumor.



**Fig. 4.** Microscopic view pilomatrixoma revealing ghost cell (arrowhead), transitional cells (large arrow) and basaloid cells (small arrow)(hematoxylin and eosin X200).

the molecular genetic department. The results showed no mutation responsible for myotonic dystrophy on chromosome 19.

During the 3-month follow-up, no recurrences were evident in the same area after surgery.

## DISCUSSION

Pilomatrixoma (also known as, calcifying epithelioma, benign calcifying epithelioma of Malherbe, tricholemmal cyst and pilomatricoma) was initially described by Malherbe and Chenantais in 1880,<sup>1</sup> who hypothesized that lesion originated from a sebaceous gland. In 1961, Forbis and Helwig<sup>2</sup> reviewed 228 such tumors with his-

tochemistry and electron microscopy and established the outer root sheath cell of the hair follicle as the cell of origin.

In 1973, Moehlenbeck<sup>3</sup> reviewed 140,000 skin tumors and noted that pilomatrixoma represented 0.12% of cases. Although pilomatrixoma can be found in any age group of patients, it occurs most often in children and young adults. A female preponderance has been reported, with a female:male ratio of 3:2. Furthermore, the majority of patients in the literature are Caucasian.<sup>3</sup> The lesion primarily affects children and adolescents, with 40% of cases occurring before age 10 and 60% before age 20.<sup>5</sup> The greatest incidence of this tumor is found in patients between 8 and 13 years of age.

The diameter of a pilomatrixoma ranges from 0.5 to 3 cm in most cases.<sup>6-8</sup> Clinically, these lesions usually appear as a firm, solitary, painless, slow-growing nodule, often with discoloration of the overlying skin. The tumors slide freely over the underlying layer of the skin. Pilomatrixoma develops most frequently in the head and neck region, followed by the upper extremities, the trunk, and the lower extremities.<sup>2,3</sup> In the head and neck region, the most common locations are the neck and the frontal, temporal, periorbital, and periauricular areas of the head.<sup>7,8</sup> No cases have been reported on the palms or soles, perhaps because hair does not grow on these areas.

Pilomatrixomas are usually solitary, but multiple foci have been reported in 3.5% of all cases. Familial pilomatrixomas and multiple familial pilomatrixomas are much rarer. Multiple pilomatrixomas and multiple familial pilomatrixomas have been associated with myotonic dystrophy,<sup>9</sup> Gardner syndrome,<sup>10</sup> and Rubinstein-Taybi syndrome.<sup>11</sup>

Diagnosis of pilomatrixoma can often be made solely on the basis of clinical features. Differential diagnosis of head and neck pilomatrixoma includes dermoid cysts, branchial cleft remnants, preauricular sinuses, adenopathy, sebaceous cysts, giant cell tumor, chondroma, foreign body reaction or malignant soft tissue tumors.<sup>6-8</sup> In the preauricular areas, pilomatrixomas can be particularly difficult to distinguish from tumors or inflammatory conditions originating from the superficial lobe of the parotid gland. Prior reviews have shown that the diagnostic accuracy rate in the preoperative diagnosis of pilomatrixoma ranges from 0% to

30%.<sup>5,7</sup> This may be due to the relative lack of familiarity of otolaryngologists with this condition.

Diagnostic tests and radiologic imaging studies are often unnecessary in the workup of a superficial, benign skin lesion such as pilomatrixoma. However, tests are sometimes done to rule out malignancy or to determine the depth of a lesion. Fine-needle aspiration has been described as a preoperative diagnostic method. However, without the presence of ghost cells (also known as shadow cells) in the aspirate, the diagnosis may be misleading. The relative superficial location of these tumors makes routine radiographic imaging unnecessary. Plain x-ray films have limited utility, but may detect foci of calcification. Computed tomography (CT) or magnetic resonance imaging (MRI) may be considered for patients who have larger tumors or tumors in unusual location. Characteristic CT findings include sharply demarcated subcutaneous lesions with various amounts of calcifications.<sup>12</sup> MRI has revealed high-signal bands on T2-weighted images which correlate with the bands formed by basaloid cells that are evident on histologic examination.<sup>13</sup> Ultrasonography has also been described as a relatively fast and noninvasive method for estimating the depth of larger masses.<sup>8</sup>

Histologically, pilomatrixoma is a deep subepidermal tumor consisting of irregular islands of epithelial cells. The cells in the islands are arranged in a circular configuration, with nucleated basaloid cells on the periphery and enucleated shadow cells in the center. The basaloid cells exhibit deeply staining basophilic nuclei with scant cytoplasm, with a lack of distinct cell borders. The ghost cells evolve from basaloid cells, and represent dead cells that retain their cellular shape and show a central unstained area that corresponds to the lost nucleus. The transitional cells, which are localized between basaloid cells and shadow cells, were thought to represent apoptotic cells that were finally proceeding to shadow cells. Calcification is mostly seen in the ghost cell regions, with the incidence ranging from 69% to 85%.<sup>2,3,6</sup> Ossification is found in 15% to 20% of specimens, probably by conversion of fibroblasts into osteoblasts.<sup>14</sup> Foreign body giant cell inflammation can also be identified in regions where keratinized debris is abundant.<sup>12</sup> In pilomatrixoma, multiple occurrences are rare. Macroscopic and microscopic aspects of multiple pilomatrixomas are not different

from the single cases.<sup>15</sup>

Although pilomatixoma is not usually associated with any chronic disease or family history, some authors pointed out that in patients with multiple pilomatixomas occurring in a familial pattern, there is a high probability of autosomal dominant disorders including myotonic dystrophy, Gardner syndrome, and Rubinstein-Taybi syndrome. In the case study profiled in this report, there were no typical features of myotonic dystrophy such as lens opacities, frontal baldness or mild mental retardation. The most common characteristic finding of the disease, myotonic phenomenon, was not observed in our case or in her family members with pilomatixoma. Further examination with venous blood samples for DNA analysis showed negative findings for myotonic dystrophy on chromosome 19. Important dermatological findings such as hirsutism, keloid and hemangiomas of Rubinstein-Taybi syndrome were not present. There was no family history of colonic polyposis.

Malignant transformation of pilomatixoma is rare.<sup>7</sup> Pilomatix carcinoma typically occurs in the posterior neck or upper back of middle-aged males (male:female ratio of 4:1 in contrast to 2:3<sup>3</sup> for benign lesions) and, with simple local excision, the recurrence rate may be as high as 60%.<sup>16</sup> Treatment is wide local excision. Rare cases of pilomatix carcinoma with distant metastasis have been reported.<sup>17</sup>

Since spontaneous regression is never observed, the standard treatment of pilomatixoma is complete surgical excision. Wide excisions with margins of 1 to 2 cm are recommended for malignant variants to minimize the risk of local recurrence.<sup>12</sup> Occasionally, the overlying skin should be excised due to the adherence of the tumor to the dermis. Recurrences after surgery are rare, with an incidence of 0% to 3%.<sup>2,6-8,12</sup> Pilomatix carcinoma should be suspected in cases with repeated local recurrences.

In conclusion, otolaryngologists should be familiar with this condition and consider it in cases of a superficial mass in the head and neck region. The clinicopathological findings of our case support the possibility that pilomatixoma could be hereditary and present without evidence of another autosomal dominant disease.

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