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CASE REPORT

Pilomatrixoma of the head and neck in children

Mohammed El-Romyssa, Jakubikova J, Pavlovcinova G

Pediatric Otorhinolaryngology Department of Medical Faculty of Comenius University, Bratislava, Slovakia. **elromyssa2@yahoo.com**

Abstract: Pilomatrixoma is a slow-growing hard mass found beneath the skin. It is most common on the face and neck but sometimes it can be found on the scalp, eyelids and arms. Most cases of pilomatrixoma occur in children under the age of seven and the condition is twice as common in females as in males. The treatment consists of surgical excision. Six patients with confirmed histopathologic diagnosis of pilomatrixoma involving the head and neck areas are reported. Presenting signs and symptoms, lesion characteristics, and treatment rendered are described. The authors review the literature and discuss the guidelines for the diagnosis and surgical management of pilomatrixoma involving the head and neck in children *(Fig. 5, Ref. 24)*. Full Text in free PDF *www.bmj.sk*.
Key words: pilomatrixoma, pilomatricoma, calcifying epithelioma of Malherbe.

Pilomatrixoma is an uncommon and harmless skin lesion derived from hair matrix cells. It is also called 'pilomatricoma', and sometimes known as 'calcifying epithelioma of Malherbe'. Calcifying epithelioma was originally described in 1880 by Malherbe and Chenanlais as a neoplasm of sebaceous glands. The term pilomatrixoma was introduced based on further studies performed in 1961 by Forbis and Helwig who demonstrated that the cells differentiated in the direction of cortical cells of the hair follicle (1, 2, 3).

Pilomatrixoma is most often diagnosed in young children but it may also affect the adults. The female-to-male ratio is nearly 3 : 2. It arises as a single skin-colored or purplish lesion on the head or neck but it may occur on any site. It is characterized by calcification within the lesion, which makes it feel hard and bony and often results in an angulated shape (the 'tent' sign) (4).

Pilomatrixomas are neoplasms of the hair cortical cells. The characteristic histological features of such neoplasms are the presence of two cell populations, namely the enucleated shadow cells and the basaloid cells lacking the nuclear features of malignancy. Calcifying deposits lacking the lamellations also help in establishing the definitive diagnosis of this benign neoplasm (5, 6).

The cause is unknown. However, recently some genetic changes have been found in the affected hair cells. Investigators have shown that at least 75 % of examined persons with pilomatrixomas have mutations in the gene *CTNNB1;* these data directly implicate beta-catenin/LEF misregulation as the major cause of tumorigenesis of hair matrix celsl in humans (7).

Pediatric ORL Department Medical Faculty of Commenius University and Children´s University Hospital. Bratislava, Slovakia

Address for correspondence: El-Romyssa Mohammed, MD, Pediatric ORL Department Medical Faculty of Commenius University and Children´s University Hospital, Limbova 1, SK-833 40 Bratislava, Slovakia. Phone: +421.0902531826

Pilomatrixomas are frequently misdiagnosed and/or missed in the differential diagnosis. Their surgical removal is curative. Recurrence after complete local excision is rare (8).

The diagnosis of pilomatrixoma can be made clinically if the characteristics of the tumor are known. Diagnostic tests and imaging studies are often unnecessary in the workup of a superficial benign skin lesion such as pilomatrixoma. However, tests are sometimes done to exclude the diagnosis of malignancy or to determine the depth of lesion. Fink and Berkowitz found ultrasound (USG) to be particularly helpful in children (9).

The characteristic sonographic appearance of pilomatrixoma is an ovoid complex mass showing an echogeneic center surrounded by a hypoechoic rim with acoustic shadowing found at the junction of dermis and subcutaneous fat and with focal thinning of the overlying dermis. Ultrasound also shows calcification. This method does not require sedation or anesthesia (10). Computed tomography and magnetic resonance imaging will add greater details to the surrounding structures and depth of the lesion, but these imaging techniques are costly and may require sedation or the use of anesthesia in children (11).

Fine-needle aspiration may reveal the presence of ghost cells, basaloid cells, and/or calcium deposition in the mass, which are diagnostic findings of pilomatrixoma. However, without the presence of ghost cells in the aspirate, the diagnosis may be misleading (12).

The differential diagnosis of pilomatrixoma is varied. Pilomatrixoma should be differentiated from epidermal and dermoid cysts. Epidermal cysts are firm, round and mobile, and their overlying skin is normal. They also present in older age groups, i.e. in adolescents and adults. Dermoid cysts are firmly attached to the underlying tissue and are often found in children. Neither of these lesions presents with irregular nodules on the skin as seen in pilomatrixoma (13).

In pilomatrixoma, multiple occurrences are rare. Macroscopic and microscopic aspects of multiple pilomatrixomas are not dif-

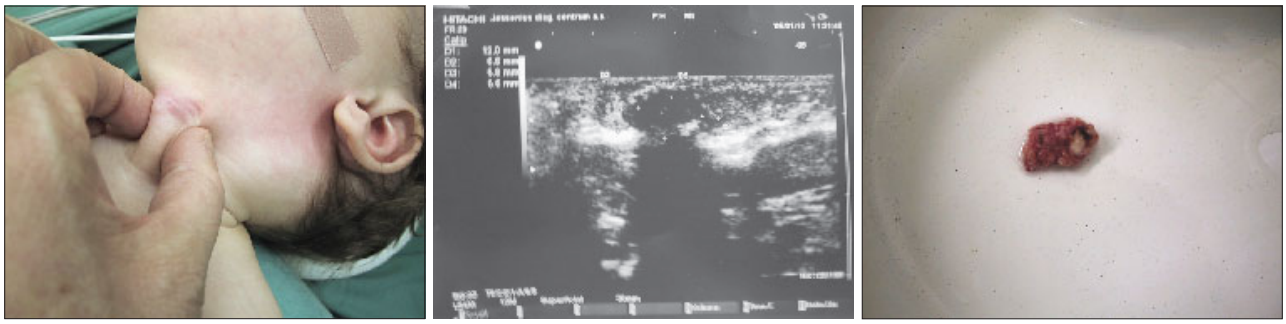

Fig. 1. a) A firm mobile lesion on the anterior aspect of the neck. b) Ultrasonography of the neck showed two subcutaneous masses. c) The removed lesion with resection of overlying skin.

ferent from the single cases. Malignant transformation of pilomatrixoma is rare (14). Pilomatrix carcinoma typically occurs in the posterior neck or upper back of middle-aged males (male-to-female ratio of 4 : 1 in contrast to 2 : 33 for benign lesions) and, with simple local excision, the recurrence rate may be as high as 60 % (15). The treatment is based on wide local excision. Rare cases of pilomatrix carcinoma with distant metastasis have been reported (16).

Case reports

Retrospectively from 2005 to 2009, six cases of patients with a confirmed histopathologic diagnosis of pilomatrixoma involving the head and neck area are reported.

Case 1

Seven-month-old female presented with a mass on the anterior aspect of the neck (under the chin). The first appearance was at birth when it was 0.5 cm in diameter and over a period of six months, it was gradually growing until it reached the size of 1.5 x 1 cm. The lesion was firm, mobile and hard. The overlying skin was normal (Fig. 1a). Ultrasonography showed two subcutaneous masses attached to each other, while one of them had cystic consistency and the other one was solid (Fig. 1b). The lesion was treated with surgical resection while the overlying skin was resected secondary to tumor adherence to the dermis (Fig. 1c).

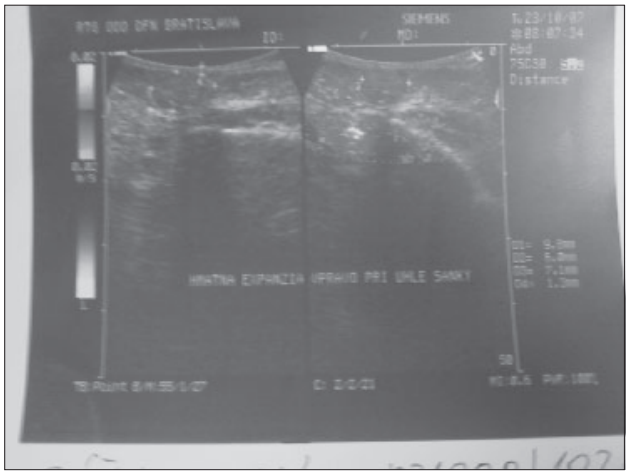

Fig. 2. Ultrasonography: subcutaneousmass with microcalcifications.

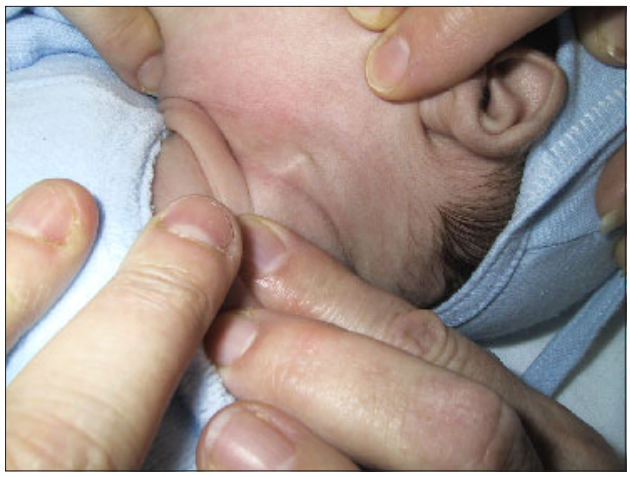

Fig. 3. A firm mass in the lateral of the neck.

Case 2

Five-year-old male presented with a submandibular mass sized 1cm in diameter. The mass gradually increased in size. It was slightly tender, firm and mobile. The overlying skin was discolored (purple-blue). Ultrasonography revealed a subcutaneous mass lying in the right submandibular aspect, and there were some microcalcifications (Fig. 2). The lesion was treated with surgical resection.

Case 3

Three-month-old male presented with a firm and mobile mass in the lateral wall of the neck with normal overlying skin (Fig. 3). Ultrasonography revealed a small solid subcutaneous mass in the left lateral wall of the neck. The lesion was treated with surgical resection.

Case 4

Four-year-old female, presented with a firm and mobile subcutaneous nodule of 0.5-cm in diameter, situated in the left preau-

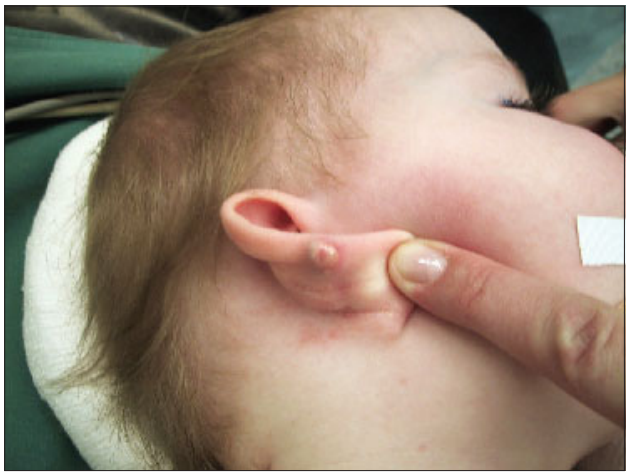

Fig. 4. A nodule in the posteriorwall of the helix of the right auricle.

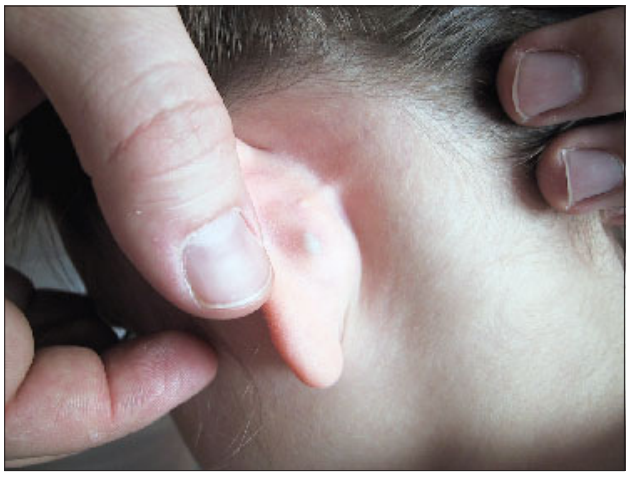

Fig. 5. A subcutaneous nodule onthe posterior wall of the left auricle.

ricular area. The overlying skin was normal. Ultrasonography revealed a well-circumscribed subcutaneous lesion sized 4 x 3 x 2 mm in the left preauricular area. The lesion was treated with surgical resection.

Case 5

One-year-old female presented with a nodule in the posterior wall of the helix of right auricle. The nodule was 1 x 1 cm in diameter and gradually increased in size. It was firm, not tender and the overlying skin was normal (Fig. 4). Ultrasonography revealed a subcutaneous mass sized 0.75 x 0.6 x 0.55 cm lying in the right auricular helix. The lesion was treated with surgical resection.

Case 6

Four-year-old female presented with a firm and mobile subcutaneous nodule in the posterior wall of the left auricle. It was 0.5 cm in size and the overlying skin exhibited bluish discoloration (Fig. 5). Ultrasonography revealed a subcutaneous mass sized 5.9 x 6.6 x 2.3 mm, strongly adhering to the cartilage of the left auricle. The lesion was treated with surgical resection.

Discussion

The pilomatrixoma tumor commonly occurs in the head and neck regions of children. Our age of presentation was similar to that in literature, with most patients presenting at age younger than seven years (13).

The diagnosis of pilomatrixoma can be made clinically. Danielson-Cohen et al (17) said that the preoperative diagnosis might be improved by being aware of the fact that pilomatrixoma was a harmless benign skin tumor in children. Patients usually present with a solitary nodule that has been slowly growing over several months or years. Patients are usually asymptomatic but some report pain during episodes of inflammation or ulceration. Pilomatrixoma lesions slide freely over the underlying area. Graham and Merwin described the "tent sign," elicited by stretching the skin over the pilomatrixoma tumor to feel the irregular surface of the mass. There is no associated lymphadenopathy. A blue discoloration may be seen (18). Yoshimura et al (19) suggested that the diagnosis of pilomatrixoma should be suspected when the mass is adherent to the skin but not fixed to the underlying tissue.

We have noticed that the most common sites of pilomatrixoma are the neck and ear areas. The lesions were located on the neck in three cases, in the preauricular area in one case, and in the auricle in two cases. In all cases, the lesions were in form of a firm, mobile and not tender subcutaneous tumor. In one case, it was tender and in two cases, a blue discoloration was present.

Patients who were clinically suspected of pilomatrixomas were subdued to ultrasonography. Hughes et al (10) suggested that the sonography of suspected pilomatrixomas in children was a useful and noninvasive procedure significantly improving the accuracy of the diagnosis of pilomatrixomas.

The treatment of choice and standard therapy of benign pilomatrixoma is based on a complete surgical resection. Occasionally, the overlying skin will need to be excised secondary to tumor adherence to the dermis as in one of our cases (Fig. 3). Morales and McGoey have advocated an incision and curettement for cosmetic preservation in large tumors or in those localized in exposed areas (20). No recurrence was found in our series.

Pilomatrixomas are frequently misdiagnosed when the evaluation is based on a clinical or radiographic examination alone. Therefore, the final diagnosis requires histopathologic examinations (21).

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 and lack of distinct cell borders. The ghost cells evolve from basaloid cells and represent dead cells retaining their cellular shape and showing a central unstained area corresponding 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. Calcifications are mostly seen in the

ghost cell regions, probably by conversion of fibroblasts into osteoblasts (23). Foreign body and giant cell inflammation can also be identified in regions where keratinized debris is abundant (24). In our patients, histologic examination following surgical excision confirmed the diagnosis; all histological reports emphasize the characteristic features of pilomatrixoma while microcalcifications were found in one case of our patients.

Conclusions

Pilomatrixoma is a rare benign skin tumor. Usually, its diagnosis is easily based on clinical findings but ultrasonography is a useful and noninvasive procedure helping to determine the depth of lesion and significantly improving the accuracy of the diagnosis of these lesions. Spontaneous regression is never observed. Complete surgical excision including the overlying skin is the treatment of choice.

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