

## Rare Clinical Variants of Pilomatricoma

*Pilomatrikomanın Nadir Klinik Varyantları*

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Pilomatricoma (calcifying epithelioma of Malherbe) is a benign adnexal tumor of hair matrix cells. Pilomatricomas are usually observed as solitary lesions. Pilomatricoma mostly makes peaks in the first and sixth decades. Female to male ratio is 3/2. More than 50% of the lesions are found on head and neck region, and 25-30% of the lesions are found on the upper extremities. The rest occurs in the trunk and rarely in the lower extremities. Pilomatricoma is not rare entity. On the contrary, multiple and perforating forms of pilomatricoma are rare entities. Although pilomatricoma is not hereditary as a rule, there are limited familial pilomatricoma cases which are published in the literature. The patients, who come to our clinic during three years and histologically diagnosed as having the pilomatricoma were analysed. Rare clinic variants of pilomatricoma that familial multiple pilomatricoma, perforating pilomatricoma are reported. In spite of the small number of the patients in our series, we believe that familial multiple pilomatricoma and perforating pilomatricoma cases will supply the literature.

**Key Words:** *pilomatricoma, familial, multiple, perforating*

Pilomatrikoma (Malherbe'nin kalsifiye epitelyoması) saç matriks hücrelerinden gelişen benign adneksal bir tümördür. Pilomatrikoma genellikle solid kitleler olarak izlenir. Sıklıkla yaşamın 1. ve 6. dekadında pik yapar. Kadın/erkek görülme oranı 3/2'dir. Lezyonların %50'den fazlası baş boyun bölgesinde, %25-30'luk kısmı üst ekstremitelerde lokalizedir. Pilomatrikoma nadir değildir. Bunun yanında ailesel multiple pilomatrikoma ve perfore pilomatrikomanın ise nadir görüldüğü bildirilmektedir. Kural olarak pilomatrikomanın herediter olmadığı söylenebilir literatürde bildirilmiş sınırlı sayıda ailesel pilomatrikoma vakası vardır. Burada 3 yıllık bir periyotta kliniğimize başvuran ve histopatolojik olarak pilomatrikoma tanısı alan 10 vaka incelendi. Literatürde nadir rastlandığı bildirilen ailesel multiple pilomatrikoma ve pilomatrikomanın nadir bir klinik varyantı olan perfore pilomatrikoma vakaları sunuldu. Bu vakalar literatür eşliğinde gözden geçirildi. Serimizdeki hasta sayısının az olmasına rağmen ailesel multiple pilomatrikoma ve perfore pilomatrikoma olgularımızın literatüre katkısı olacağını düşünüyoruz.

**Anahtar Kelimeler:** *pilomatrikoma, ailesel, multiple, perfore*

Pilomatricoma is a benign adnexal tumour of hair matrix cells. It is most frequently seen in head and neck region and upper limbs (1).

Malherbe and Chenantais defined the "calcifying epithelioma" in 1880. In the beginning they thought this as a tumor of sebaceous glands. In 1905, this mistake was corrected by Malherbe and the entity was renamed as "calcifying epithelioma of Malherbe" (2). In 1949, Lever and Griesemer (3), claimed that

the tumor was taking origin from the hair matrix cells, and 1961, it was named as "pilomatricoma by Forbis and Helwig" (4). Then, pilomatricoma was changed to pilomatricoma which is more etymologically accurate (2).

Pilomatricoma is not rare entity. When the literature is reviewed, incidence varies from one in 500 to 924 dermatologic specimens (5,6). On the contrary, multiple and perforating forms of pilomat-

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**Table 1:** Patients information. ( L: left, R: right, lat.:laterale, month: mn., year: yr.)

Case	Age (year)	Sex	Localization	Size (cm)	Duration	Clinical diagnosis	Histopathological diagnosis
1	4	M	L infraorbital	1,5x1x0,6	4 mn	Organized hematom	pilomatricoma
2	21	M	L leg	3x2x1	2 yr.	pilomatricoma	pilomatricoma
3	21	F	L right	1x0,7x0,7	18 mn	pilomatricoma	pilomatricoma
4	76	M	R eyebrow	1,5x1x0,5	3 yr.	epidermoid cyst	pilomatricoma
5	47	F	Scalp	0,5x0,5x0,3	3 yr	epidermoid cyst	pilomatricoma
6	59	M	L pectoral, L subclavicular	3x1,5x0,5 1x0,8x0,5	1 yr	epidermoid cyst	pilomatricoma
7	18	F	L eyebrow	0,5x0,5x0,3	6 mn.	epidermoid cyst	pilomatricoma
8	66	E	R temporal	0,8x0,7x0,3	6 mn.	Basalcell carcinoma	pilomatricoma
9	15	F	Anterior cervicale	2,5x1,5x1	1 mn.	epidermoid cyst	pilomatricoma
10	17	F	R lat. cantus L preauricular Posterior cervicale	4x3x1,5 0,6x0,5x0,2 2x1,5x0,5	4 yr.	pilomatricoma	pilomatricoma

ricoma are rare entities (4,7-11). Although pilomatricoma is not hereditary as a rule, there are limited familial pilomatricoma cases which are published in the literature (12-14).

In this article, we report 10 pilomatricoma cases operated in our clinic during three years and presented three of them which are rarely seen in the literature (Table-1).

## Case Reports

### Case 2

21 year old male patient presented with a 2 year history of painless mass on the left leg. Examination showed mobile, well defined,

firm, 3x2x1 cm subcutaneous nodule covered by normal skin. Local excision of nodule with overlying skin was performed (Figure 1.A-B). Histopathologic examination revealed that, tumor was consisted of island of epithelial cells. It was composed of basophilic and eosinophilic shadow cells. Areas of calcification and ossification were detection in lesion . Lesion was seperated from the neighbour skin with the regular border.

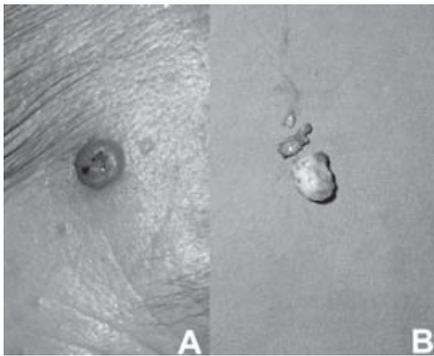
### Case 8

66 year old female patient presented to our clinic with the mass on her right temporal area. She had noticed the asymptomatic nodule with ulceration about 6 months previously. Physical examination revealed, bluish colored nodule with ul-

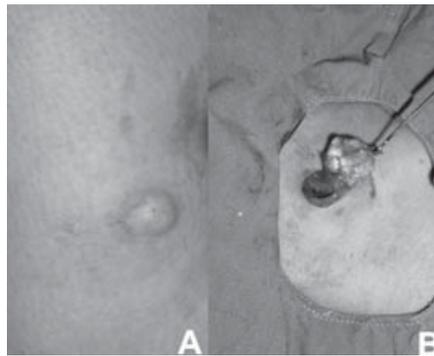
cer in center. That was 0,8x0,7x0,3 cm. size on the right temporal area. Lesion was excised by clinical diagnoses of BCC (Figure 2.A-B). On microscopic examination, shadow cell groups, basophilic cells and cystic cavity were observed. There were also foreign body giant cells. The epidermis adjacent to the tumor invaginated.

### Case 10

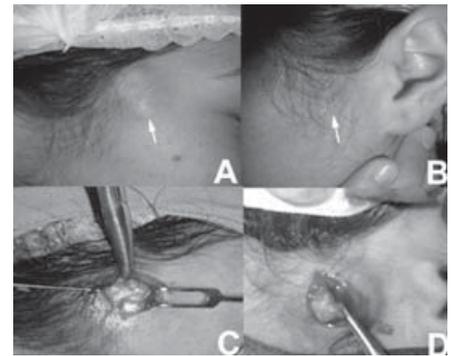
17 year old female patient referred to our clinic because of the masses in her face and neck since the age of 13 years. When she examined, the masses, which were painly, hard, mobile were found out and these masses did not cause colour change. The mass in the laterale of right eyebrow was 4x3x1,5 cm., the mass in the left preauricular



**Figure 1:** Case 2 A) Firm, nodular lesion on the leg. B) Peroperative view of the lesion



**Figure 2:** Case 8 A) A nodular lesion on the temporal region with ulceration. B) Intraoperative view of the lesion



**Figure 3:** Case 10 A-B) Preoperative lateral views of masses. C-D) Peroperative views of masses

area was 0,6x0,5x0,2 cm., and the mass in the posterior neck area was 2x10x0,5 cm. These three masses were surgically removed (Figure 3.A-C). On the histopathological observation, a few cells in the bazloid features around the lobuller occuring from the cell remnants which did not include nucleous in the fibroz stroma drew attention. In addition to this, calcification areas were found out. There were multinuclear giant cells and lenfositier inflatration were seen adjacent.

After the pathologic diagnosis, systematic illnesses, which could be seen with pilomatricoma, like myotonic distrofi, Gardner syndrome, Reynaud phenomene were examined. However, none of these pathologies were detected. Moreover, it was learned that her sister, brother and uncle had the similiar masses. Her uncle's masses in his neck and left preauciliar areas were taken out 10 years ago. Its histopatological diagnosis was pilomatricoma. A hard mass was found in his brother's right arm. Histopatological diagnoses of this mass came as pilomatricoma. Also, her sister had a painless mass. Histopatological examination of the mass could not be done because she was in abroad. These three cases are not attached to the table.

## Discussion

Pilomatricoma usually presents as firm to stone hard, asemptomatic nodule with dimensions ranging from 0,5 to 5 cm. The overlying skin is generally normal, but may show a blue- red discoloration (1).

Etiology of pilomatricoma is not known. Microscopic, biochemical enzymatic, polarischopic, and immunohistochemical properties reveals that it originates from the hair matrix cells (15). Normal hair matrix is characterized with anogen, telogen and catagen cycilus (16). This cycilus is controlled with programmed cell death (apoptosis) (17). It's thought that it is a defect occuring in progress of the normal anogen phase in pilomatricomas (18). "bcl-2", which is a proto-oncogen, suppresses the apoptosis in both a benign and malign tumor. In addition, both apoptosis and "bcl-2" are the critical factors in the normal hair follicule progressing. Farrier and his friends(17) showed that pilomatricomas are painted strongly with "bcl-2" in their immunohistochemical study. They pointed out that this raised "bcl-2" painting causes the wrong apoptosis

suppression. This contributes to the pathogenesis of pilomatricoma. In some studies it was shown that  $\beta$  catenin mutation also play a role in this pathogenesis (19,20).  $\beta$  catenin misregulation is responsible for the anormal proliferation of hair matrix cells (19).  $\beta$  catenin takes a role in cell-cell adhesion rather than celluler proliferation in pilomatricoma tumorogenesis (20).

Pilomatricoma mostly makes peaks in the first and sixth decades. %40 of the patients are in the first decade. Female to male ratio is 3/2 (3,7,9). More than %50 of the lesions are found on head and neck region, and %25-30 of the lesions are found on the upper extremities. The rest occurs in the trunk and rarely in the lower extremities (2-12). Pain and tense seen in % 20 to 35 of cases (21-22).

In our study, female to male ratio was 3/2. Except the four whose age range was between fifth and eighth decades the age of other patients were in the first and second decades. In two of our cases the masses were painfull (%20). Although in most of our cases lesions were located in the head and neck region, one of them was located in the leg (Table 1).

Pilomatricomas are usually observed as solitary lesions. 4 clinical variances of were defined; eruptif type, perfore type, myotonic distrofi which occurs with or without familial type and recurrent invaziv nonmetastatic pilomatrix carcinoma (2).

Although most of the articles report the occurrence rate of multiple lesions as %2-3,5 (2,3,7-12), in the article of Yoskimoto this was reported as %26 (23). When they compared this high rate with other broader series (24,25), they conclude that this may be due to small number of cases in their report or racial factors (23). Multiple pilomatricoma may be associated myotonic distrofi, sarkoidozis, Gardner syndrome, Turner syndrome and Reynaud phenomenon. Multiple pilomatricoma is rare without myotonic distrofi. The association of multiple pilomatricoma and myotonic distrofi is reported as %75 (2,3,9,12-14). Multiple pilomatricoma was seen in two of our patients .

The familial pilomatricoma is the most rarely seen type. Up to now, only 10 familial cases have been reported (7,11-14). Of these some were soliter and some were multiple. Four familial pilomatricoma cases associated with miyotonic distrofi were multiple (11,14). In our familial case although both our patient and her brother not have myotonic distrofi.

Perforated pilomatricoma is seen rarely. When literature is examined, it was seen that up to now, only 10 cases have been published (3,10,11). In our series, only one patient had perforated pilomatricoma.

Clinic diagnosis of pilomatricoma is difficult. The wrong diagnosis rate ranges from %71 to % 79 (21,25). Pilomatricoma can be frequently confused with epidermoid cyst, calcifying lymf node, organized hematoma, foreign body reaction, dermoid cyst or parotis gland tumours. When perforation is seen, it may be misdiagnosed as in skin cancer (2-12,21-26). In our study, one lesion was misdiagnosed as organized hematoma and one lesion was misdiagnosed as BBC. Four lesions were misdiagnosed as epidermoid cyst (Table-1).

Spontaneous regression has never been observed. The treatment of choice is surgical excision. Incomplete resections have been followed by local recurrence. After the resection local recurrence rates were about % 2-6 (4,21).

Diagnosis of pilomatricoma is usually made with histopathological examination. Lesion is usually located in the lower dermis and subcutaneous fat. It is usually surrounded by connective tissue capsule and, sharply demarcated from skin and subcutaneous. Irregularly shaped islands of either basophilic or shadow epithelial cells are seen. Ba-

sophilic cells are usually arranged periphery of the tumor islands. From periphery to the center they lose their nucleuses step by step, and in the center of island shadow cells are seen. They are dyed eozinophilic (1,18). As the lesion age the number of basophilic cells decreases (3). Calcium deposits may occur in 75 percent of lesions and ossification areas can be seen in 15-20 percent of lesions (2,3,12). When we observed the histopathologic investigation of our patients, the ossification areas were approximately %50 in our five cases.

## Conclusion

When the literature was reviewed, it was seen that familial pilomatricoma and perfore pilomatricoma, which are the clinic variances of pilomatricoma, have been informed rarely. In spite of the less number of the patients in our series, we believe that familial multiple pilomatricoma and perfore pilomatricoma cases will supply the literature.

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