CASE REPORT OLGU SUNUMU

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Periorbital Pilomatrixoma

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ABSTRACT Pilomatrixoma is a rare benign tumor originating from the hair follicle matrix, most commonly developing during childhood and adolescence. While it can arise in various parts of the body, it is predominantly observed in the head and neck region during childhood. The incidence has been found as 1/824 based on the analysis of dermatohistopathological materials in the literature. This study aims to evaluate a case series of patients diagnosed histopathologically with pilomatrixoma. All cases presented with a painless, well-circumscribed, nontender, mobile mass located under the eyebrow. Simple excision was performed in all cases without any postoperative complications. Although slowly growth is typically anticipated, rapid growth was noted in 2 of our cases. In conclusion, although pilomatrixoma is rare, it should be considered in the differential diagnosis of masses in the periocular region, particularly those arising under the eyelid and eyebrow in childhood. Surgical excision remains the gold standard treatment for this condition.

Keywords: Pilomatrixoma; orbita

ÖZET Pilomatriksoma, kıl kökü matriksinden köken alan, genellikle çocukluk ve adölesan döneminde görülen nadir ve benign bir tümördür. Vücudun birçok bölgesinde gelişebilmekle birlikte, çocukluk çağında sıklıkla baş ve boyun bölgesinde izlenir. Literatürde bildirilen dermatohistopatolojik materyallerin incelenmesi sonucunda insidansı 1/824 olarak bulunmuştur. Bu çalışmanın amacı, histopatolojik olarak pilomatriksoma tanısı almış bir olgu serisinin değerlendirilmesidir. Olgu serisindeki tüm hastalar, kaş altında, ağrısız, iyi sınırlı, hassasiyet göstermeyen ve mobil kitle ile başvurmuştur. Tüm olgularda postoperatif komplikasyon gelişmeksizin basit eksizyon uygulanmıştır. Literatürde olgularda yavaş büyüme bildirilmesine rağmen, 2 olgumuzda hızlı büyüme izlenmiştir. Sonuç olarak, pilomatriksoma nadir görülmekle birlikte, çocukluk çağında perioküler bölgede, özellikle göz kapağı ve kaş altında ortaya çıkan kitlelerin ayırıcı tanısında mutlaka göz önünde bulundurulmalıdır. Cerrahi eksizyon, bu tümörün tedavisinde genellikle altın standart yöntemdir.

Anahtar Kelimeler: Pilomatriksoma; orbita

Pilomatrixoma, also referred to as Malherbe's calcified epithelioma, is a benign tumor originating from the matrix cells of hair follicles.[1] It was first described by Malherbe and Chenantais in 1880, with the term "pilomatrixoma" later introduced by Forbis and Helwig in 1961.[2,3]

Pilomatrixoma is more commonly observed during childhood and adolescence. While it can develop in various parts of the body, it is relatively more prevalent in the head and neck region. It is considered a rare lesion of periocular tissues. Due to the lack of preoperative suspicion, pilomatrixoma is often clinically misdiagnosed and overlooked in the differential diagnosis. A definitive diagnosis is typically established only after surgical excision and histopathological evaluation.[1]

The aim of this study is to present cases of histopathologically confirmed pilomatrixoma in pa-

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tients presenting with a mass located within the periorbital region.

CASE REPORTS

CASE 1

An 8-year-old female patient presented to our clinic with a complaint of a painless mass under the left eyebrow, which had been steadily growing over the past 4 months. On ophthalmologic examination, her visual acuity was 20/20 with a refractive correction of -1.25 D in the right eye and -1.0 D in the left eye. The anterior and posterior segment examinations, pupillary light reflexes, and ocular motility were all normal in both eyes. A well-circumscribed, nontender, mobile subcutaneous mass measuring 6×6 mm was identified under the left eyebrow. The overlying skin showed no discoloration or other abnormalities. The patient underwent a total excisional biopsy under general anesthesia. The mass was completely excised through a linear incision made just below the eyebrow.

Macroscopically, the excised mass measured 6×6×5 mm, appeared white with brownish areas on the surface, and exhibited a well-circumscribed, multilobular structure (Figure 1). On cross-sectional examination, the lesion displayed a homogeneous cream-white solid appearance. Microscopically, the specimen revealed a hyperchromatic basaloid cell layer, ghost cells with indistinct nuclei, and surrounding histiocytic cells (Figure 2). Histopathological findings confirmed the diagnosis of pilomatrixoma.

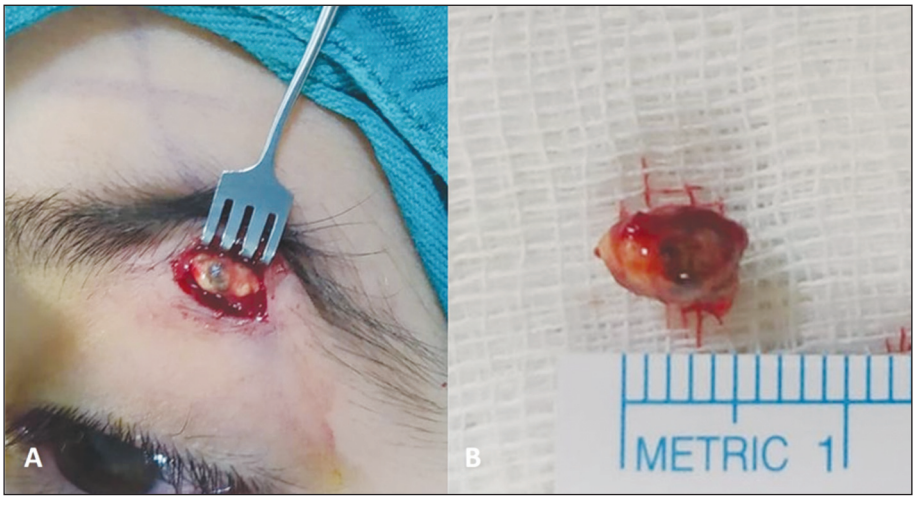

FIGURE 1: Macroscopic view of periocular pilomatrixoma. **A)** A well-circumscribed mass with a left under-brow incision. **B)** 6x6x5 mm tumoral mass removed under the left eyebrow

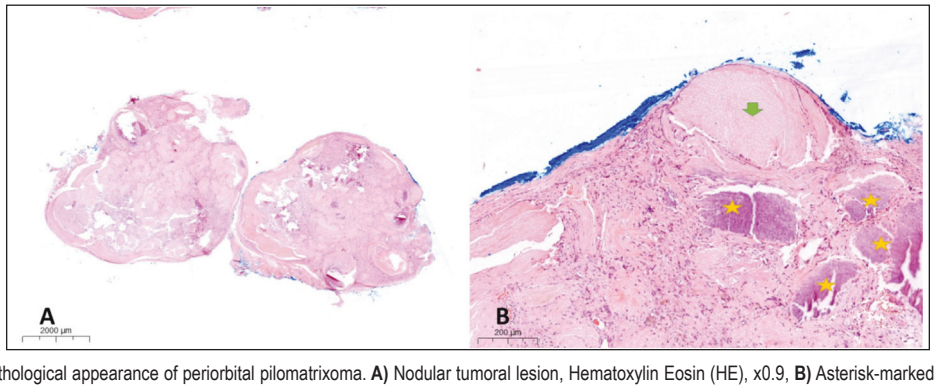

FIGURE 2: Histopathological appearance of periorbital pilomatrixoma. **A)** Nodular tumoral lesion, Hematoxylin Eosin (HE), x0.9, **B)** Asterisk-marked hyperchromatic basaloid cell layer and arrow-indicated ghost cells observed as a layer with indistinct nuclei, accompanied by histiocytic inflammatory response surrounding them. HE, x8.4

CASE 2

A 5-year-old female patient presented to our clinic with a complaint of a painless, reddish mass under the left eyebrow that had been steadily growing over the past month. Ophthalmologic examination revealed a visual acuity of 20/20 in both eyes with a refractive correction of +0.50 D. The anterior and posterior segment examinations, pupillary light reflexes, and ocular motility were normal bilaterally. A well-circumscribed, nontender, mobile subcutaneous mass measuring 5×5 mm was identified under the left eyebrow, with visible subcutaneous telangiectatic vessels. The overlying skin showed a pink to purple discoloration (Figure 3). The patient underwent a total excisional biopsy under general anesthesia and the mass was surgically excised en bloc through a linear incision made below the eyebrow.

Macroscopically, the excised mass measured 11×6×7 mm, and its cross-sectional surface displayed a homogeneous cream-white solid appearance. Histopathological evaluation confirmed the diagnosis of pilomatrixoma.

CASE 3

A 6-year-old female patient presented to our clinic with a complaint of a painless mass under the left eyebrow that had been steadily growing for approximately 4 years. On ocular examination, her visual acuity was 20/20 in both eyes. The anterior and posterior segment examinations, pupillary light reflexes, and ocular motility were normal bilaterally. A well-circumscribed, nontender, mobile subcutaneous mass measuring 5×6 mm was identified under the left eyebrow. No discoloration or abnormalities were observed on the overlying skin. The patient underwent a total excisional biopsy under general anesthesia and the mass was excised en bloc through a linear incision under the eyebrow.

FIGURE 3: A painless, red-blue mass under the left eyebrow that has been growing steadily for about 1 month

Macroscopically, the excised mass measured 5×5×3 mm and exhibited a nodular appearance. Histopathological evaluation confirmed the diagnosis of pilomatrixoma.

Written informed consent for inclusion in this case series was obtained from the parents of all patients.

DISCUSSION

Pilomatrixoma typically presents as a solitary lesion, predominantly affecting younger individuals. It is estimated that approximately 40% of cases occur during the first decade of life, while an additional 20% develop during the second decade.[4] In some studies, the incidence of pilomatrixoma has been reported to range between 0.001% and 0.0031%.[1,4,5] It shows equal incidence in both males and females.[6,7] The head and neck region is the most commonly affected site for pilomatrixoma, although it can also occur in the upper extremities, trunk, and lower extremities.[4] In a study analyzing 95 pediatric histopathologically diagnosed with pilomatrixoma over 20 years, 12% had lesions in the periorbital region.[6] Another single-center study found periorbital involvement in 17% of 150 diagnosed cases with pilomatrixoma.[8]

Clinically, pilomatrixoma presents as a well-circumscribed, mobile subcutaneous mass, often with red-blue discoloration and a firm to lumpy consistency on palpation. Several studies have documented its clinical characteristics, including onset during childhood or young adulthood, an average size of 10 mm or smaller, a firm-cystic consistency, slow growth over weeks to months, a pink to purple coloration with yellowish areas, and the presence of subcutaneous telangiectatic vessels.[9]

Upon closer examination, pilomatrixoma reveals numerous islands of epithelial cells composed of basophilic cells arranged at the periphery and ghost cells located centrally. As the tumor progresses, basophilic cells appear to gradually lose their nuclei, transforming into ghost cells. Calcification is reported in approximately 75% of cases. In necrotic regions, dense layers of eosinophilic keratinous material may develop, potentially triggering a foreign body giant cell reaction.[7]

Preoperative diagnosis of pilomatrixoma is often challenging, with definitive diagnosis typically relying on histopathological evaluation following surgical excision.[6,8] The most common preoperative misdiagnoses for pilomatrixoma are epidermoid and dermoid cysts.[6,10] Epidermoid cysts, more common in adults but occasionally seen in children, range from 1 to 5 cm, are mobile, painless, and covered by intact epidermis. Unlike pilomatrixomas, they exhibit a diffuse homogeneous yellow color when filled with keratin and are softer on palpation. Dermoid cysts, primarily affecting children and young adults, are typically attached to the orbital rim, with overlying skin that remains mobile and normal in appearance. In contrast, pilomatrixomas are mobile within subcutaneous tissue but adherent to the overlying, often discolored skin, and have a characteristic rock-hard consistency, distinguishing them from dermoid cysts.[6,10] Al-Muhaylib et al. reported a 3-year-old case of pilomatrixoma that was initially suspected to be a dermoid cyst. However, following excision, the lesion was histopathologically confirmed as pilomatrixoma.[11]

Treatment involves total surgical excision with approaches varying by lesion size and location. For lesions in the eyebrow region, the under-brow approach, which involves a small incision, is particularly effective. In cases of larger lesions, an upper eyelid crease incision may be preferred.[12]

While pilomatrixomas generally grow slowly over months to years, 2 cases in our study exhibited rapid enlargement within 1-4 months. This is an uncommon but significant feature of pilomatrixoma, distinguishing it from its typical presentation of slow, gradual enlargement. A case of a rapidly growing eyelid mass diagnosed as pilomatrixoma, with enlargement occurring within two months, has been reported in the literature.[13] Additionally, rapidly growing pilomatrix carcinomas have also been described.[14] Though rare, malignant transformation can occur, and incomplete excision may lead to metastasis to the bones, lungs, and lymph nodes.[15]

In conclusion, pilomatrixoma should be considered in the differential diagnosis of periorbital masses, even in cases with atypical or rapid growth. Definitive diagnosis necessitates excision and histopathological confirmation. Prompt recognition and surgical management are critical for optimal patient outcomes, particularly in atypical presentations.

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal;* ***Design:*** *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal;* ***Control/Supervision:*** *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal;* ***Data Collection and/or Processing:*** *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal, Bilge Ayça Kırmızı, Elif Tuncel;* ***Analysis and/or Interpretation:*** *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal, Bilge Ayça Kırmızı, Elif Tuncel;* ***Literature Review:*** *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal;* ***Writing the Article:*** *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal;* ***Critical Review:*** *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal;* ***Materials:*** *Pınar Aydın Ellialtıoğlu, Melek Banu Hoşal, Bilge Ayça Kırmızı, Elif Tuncel.*

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