A Case of Vaginal Intraoitus Localised Pilomatrixoma

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Abstract

Pilomatrixoma is a rare and usually benign tumour arising from the hair follicle. It usually presents as painless subcutaneous nodules on the head, neck and upper extremities in children and young adults. Pilomatrixoma may occasionally occur in atypical locations and can cause diagnostic difficulties due to its non-specific clinical presentation. In this case report, a 36-

year-old woman presented with a painless palpable mass localised to the vaginal introitus and inability to engage in coitus. After total surgical excision, no recurrence was detected in a one-year follow-up. Since its initial report in this localization, it has been thought to contribute to the literature by raising awareness in the differential diagnosis of vulvar and vaginal masses, particularly in gynaecological practice and by guiding other specialties that may encounter similar cases.

Introduction

Pilomatrixoma is defined as a slow-growing, painless, benign tumour that develops from the skin and subcutaneous tissue. It was first described by Malherbe and Chenantais in 1880 and was believed to originate from the sebaceous glands. However, it is now known that this tumor originates from hair follicle matrix cells.

It is reported more frequently in women and is more common in areas such as the head, neck and upper extremities. Rarely, it may also occur on the trunk and lower extremities [2]. It usually presents as a single subcutaneous nodule, while cases of multiple pilomatrixomas are very rare. Pilomatrixoma, which frequently occurs in children and young adults, can also be seen in older age groups in rare cases. Although the exact incidence and prevalence are unknown, it is considered a rare tumor. Although large-scale epidemiological studies are not available, one study reported an incidence of 1-3 cases per 1,000 dermatohistopathological samples examined. Some studies have reported that 50% of cases are diagnosed before the age of 20 [3].

The treatment is total surgical excision. Local recurrence may rarely occur. Although it is known as a benign tumor, cases of pilomatrixocarcinoma, the malignant form of pilomatrixoma, have also been reported in the literature. However, the possibility of malignant transformation is not known. It is stated that malignant forms exhibit a more aggressive biological behavior than benign forms and have a high potential for local recurrence and metastasis. [4,5]

Pilomatrixoma cases can be easily confused with benign or malignant lesions. Especially in the case of atypical localisation, it may make it difficult for the clinician to consider and diagnose in the differential diagnosis in the preoperative period. In such cases, histopathological examination is the gold standard method for definitive diagnosis.

In the literature, only a few cases of pilomatrixoma in the vulvar region have been reported, and no case of pilomatrixoma in the vaginal introitus has been reported. This case report describes the surgical excision of a pilomatrixoma tumour located in the vaginal introitus, blocking the vaginal entrance and preventing coitus in a 36-year-old woman, the results of

histopathological examination and the first report of this rare localisation in the literature. With this case report, we aim to raise awareness about this rare tumor and its localization, and to contribute to the diagnosis and treatment approaches, especially in gynecology practice and other specialties.

Case Report

A 36-year-old female patient presented with a painless mass in the genital area, which was palpable, had been gradually growing for the last 6 months, and interfered with coitus. There was no known disease or trauma history in her medical history. On physical examination, a superficial, hard, partially mobile, smoothly circumscribed mass with a diameter of approximately 4.5 cm, covering the vaginal introitus and covered with vaginal mucosa, was detected. The overlying mucosa appeared normal and no inflammatory findings were found (Figure 1).

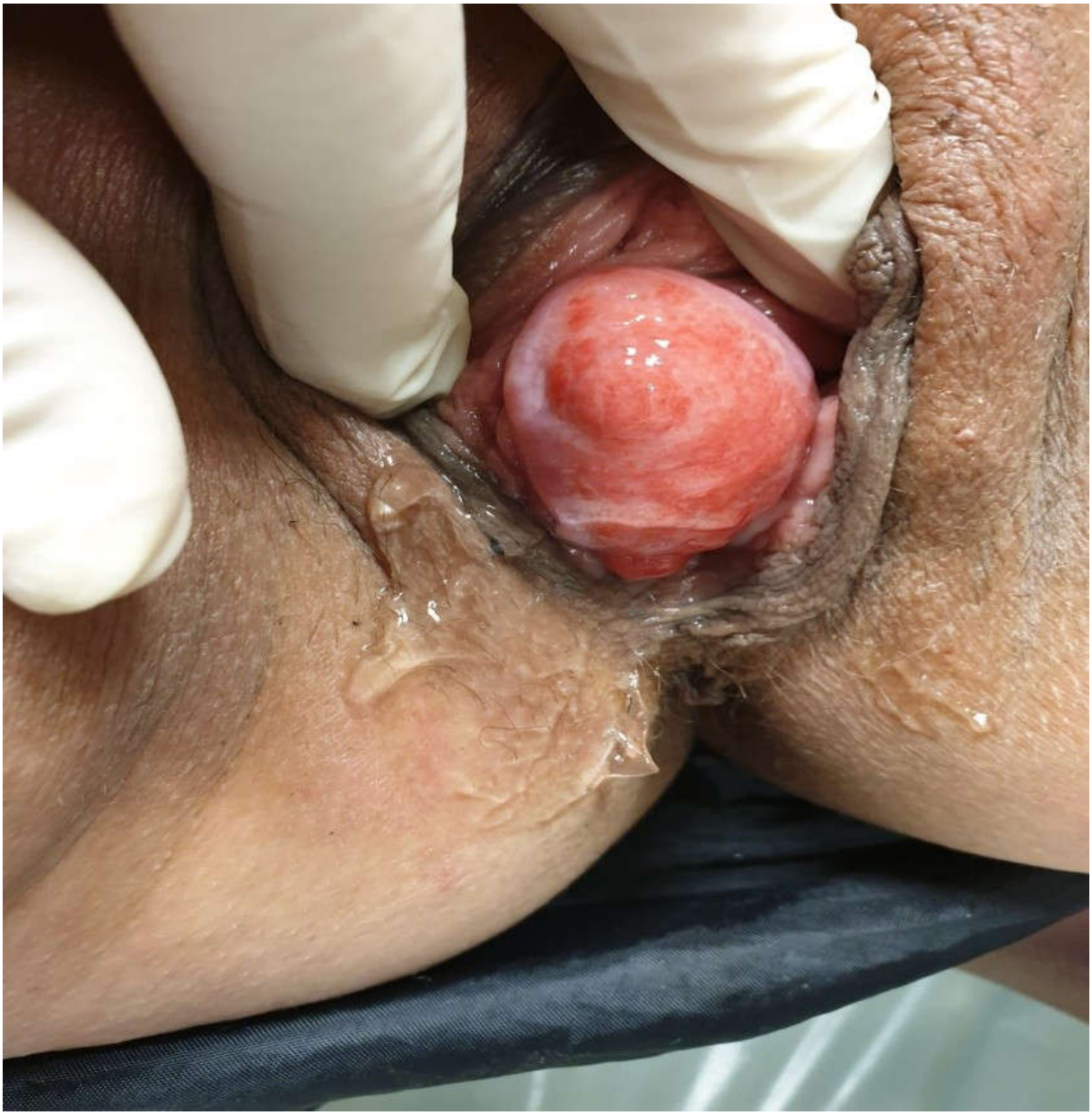

Figure 1. Pilomatrixoma tumor obstructing the vaginal introitus.

Ultrasonography confirmed that the mass was subcutaneously localised and separated from other anatomical structures. Epidermal cyst, lipoma, teratoma and vulvar malignancy were considered among the clinical prediagnoses.

Total surgical excision was performed under anaesthesia. During the operation, it was observed that the tumour did not invade the surrounding tissue and the excised mass was removed entirely. Bleeding was controlled and the incision was closed with sutures (Figure 2). There were no complications during and after the procedure.

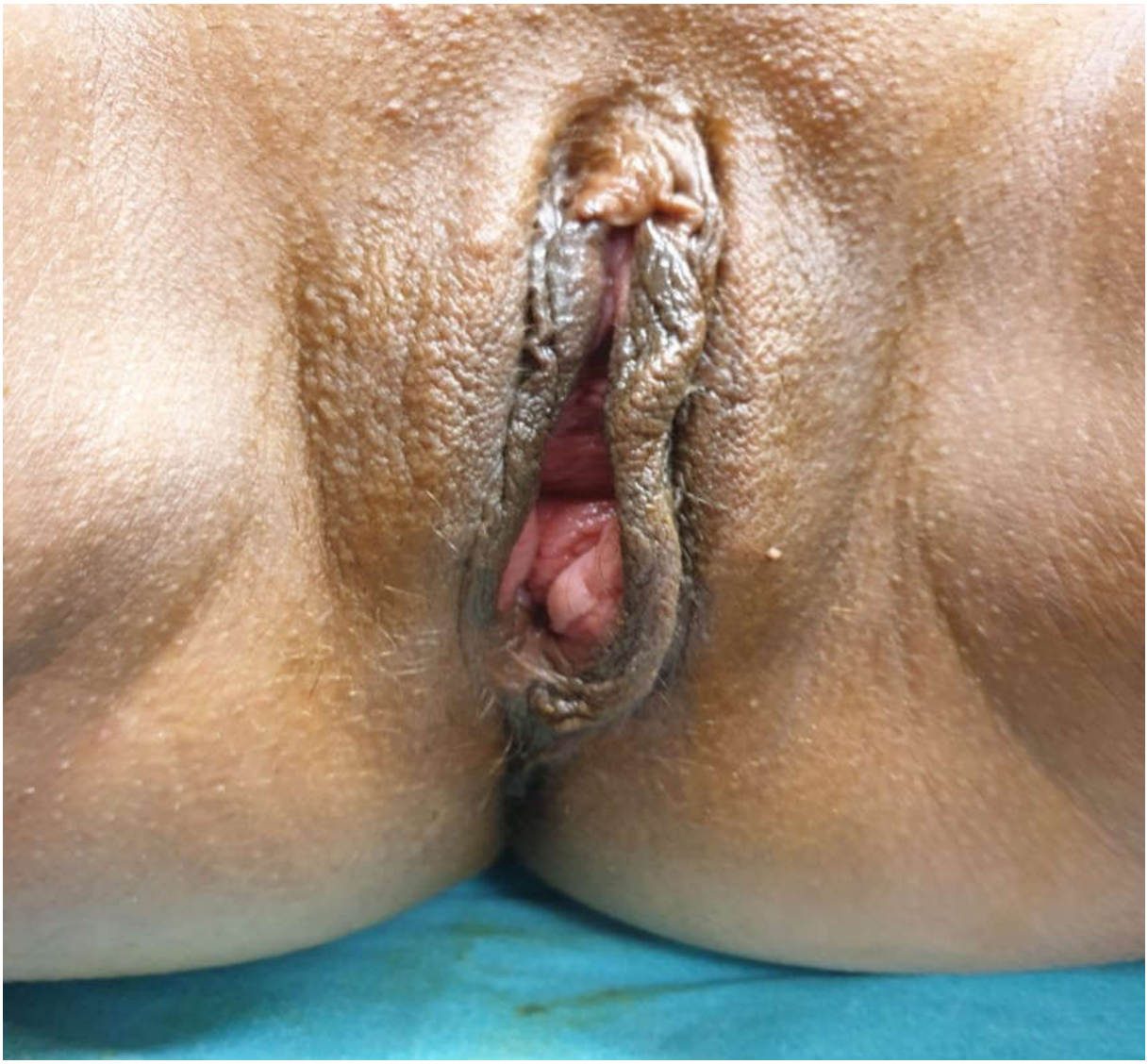

Figure 2. Postoperative appearance.

On pathological macroscopic examination, the tumour was defined as well-circumscribed, elastic, covered with mucosal tissue, cream-white in cross-section and 4.5x3.5x3 cm in size. In microscopic analysis, the tumor was covered with multilayered squamous epithelium and consisted of matrixial cells and ghost cells, with good demarcation from the surrounding tissue under the epithelium. It contained sebaceous material in the lumen and granulomatous inflammation foci around it. Mitotic activity was not detected and no findings compatible with malignancy were found. The final diagnosis was reported as 'Benign Pilomatrixoma (Malharbe Tumour)'.

In this case report, the patient was informed in detail that their case could be published in a scientific study and provided written consent for the use of their images. The study was conducted by the Declaration of Helsinki.

Discussion

Pilomatrixoma is a benign tumour originating from the matrix of the hair follicle. They are usually superficial, smooth, contoured neoplasms. It is also known as Malharbe's calcified epithelioma (Malharbe's tumour) [6]. These tumors, which are frequently seen in childhood and young adulthood, are usually localized in the head, neck and upper extremities. In the vulvar region, cases of pilomatrixoma are rarely reported. In one of the case reports in the literature, it was reported that the tumour was located in the labium majus [7], and the other 2 cases were reported to be located in the vulva without specifying the specific location [8]. Our 36-year-old case is the first reported in the literature in terms of both being located in the vaginal introitus, a rare anatomical localization, and preventing coitus by closing the vaginal introitus. This situation is especially important for gynaecological practice.

In the preoperative period, it may be confused with other pathologies, such as Bartholin abscess, epidermal cyst, lipoma, teratoma, or malignant tumors, especially in the vulvar and vaginal regions. This case is a rare example with a difference in localisation and may lead to the need to review gynaecological diagnostic algorithms, preoperative evaluation and treatment protocols. In specific localisations such as vaginal introitus, it is critical to consider the preliminary diagnosis of pilomatrixoma in the differential diagnosis in terms of treatment planning and patient management.

The physical examination findings and ultrasonography evaluation performed in the preoperative period helped us to define the characteristics of the tumour, and total surgical excision was planned considering benign causes. Total surgical excision is generally accepted as an adequate treatment method for such tumours [6,9]. Histopathological evaluation is the gold standard for definitive diagnosis. However, since recurrence (2-3%) or transformation to malignancy is rarely reported, detailed histopathological examination of the excised material and complete evaluation of the surgical margin are critical [10]. Since pilomatrixoma has a benign character, the prognosis after treatment is generally excellent. Since recurrences are caused by inadequate removal of the tumour during surgical excision, total excision and tumour-free surgical margins are of great importance. In this case, the tumour was completely excised and no recurrence or complications were observed during the 1-year follow-up.

This case report emphasises the diversity of pilomatrixoma in terms of both anatomical location and clinical presentation. We think that this case, which closes the vaginal introitus, causing the inability to have intercourse and is reported for the first time in this anatomical location, will increase the awareness of clinicians about these rare lesions. The knowledge that pilomatrixoma cases may occur in different localizations should not be forgotten in the

differential diagnosis of vulvar and vaginal masses, especially in gynaecology practice. Total excision without radical surgery is curable, and morbidity can be reduced, which will add a new dimension to gynaecology practice.

Conclusion

This case demonstrated that pilomatrixoma tumors do not always exhibit typical features and may present with unexpected clinical presentations in rare cases. The limitation of our case report is that it is based on a single case. Therefore, further studies should be performed to better understand the clinical features and treatment approaches of rare pilomatrixoma tumours. This will also contribute to the lack of prevalence and incidence data. Reporting this case will increase clinical awareness of subcutaneous masses in the genital area and will help in future patient management. Consideration of such rare tumors by physicians, especially in gynecology practice, during their diagnostic process may give patients a chance for early diagnosis and appropriate treatment. Additionally, it will be helpful in guiding other specialties that may encounter similar cases.

Conflict of interest

None declared

Informed consent of the patient

In this case report, the patient was informed in detail that their case could be published in a scientific study and provided written consent for the use of their images. The study was conducted by the Declaration of Helsinki.

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References

1. Jones CD, Ho W, Robertson BF, Gunn E, Morley S. Pilomatrixoma: a comprehensive review of the literature. Am J Dermatopathol. 2018, 40:631-41. 10.1097/DAD.0000000000001118

2. Migirov, L., Fridman, E., & Talmi, Y. P. Pilomatrixoma of the retroauricular area and arm. *Journal of Pediatric Surgery*. 2002, *37*(8), 1-2.

3. Çil, Y., & Çil, H. Baş Boyun Bölgesinde Pilomatriksoma: Olgu Sunumu. *Eskisehir Medical Journal*. 2021, *2*(1), 40-42.

4. Jones, C., Twoon, M., Ho, W., Portelli, M., Robertson, B. F., & Anderson, W. Pilomatrix carcinoma: 12 year experience and review of the literature. *Journal of Cutaneous Pathology*. 2018, *45*(1), 33-38.

5. Papadakis, M., De Bree, E., Floros, N., Giannikaki, E., Xekalou, A., & Manios, A. Pilomatrix carcinoma: more malignant biological behavior than was considered in the past. *Molecular and clinical oncology*. 2017, *6*(3), 415-418.

6. Zhao A, Kedarisetty S, Arriola AG, Isaacson G: Pilomatrixoma and its Imitators. Ear Nose Throat J. 2024, 103:183-9. 10.1177/01455613211044778).

7. Öcal, D. F., Türkyılmaz, E., Gültekin, İ. B., Çekmez, Y., & Günçe, S. Pilomatrixoma localized in vulva. *Medeniyet Medical Journal*. 2016, *31*(4).

8. Baker GM, Selim MA, Hoang MP. Vulvar adnexal lesions: a 32-year, single-institution review from Massachusetts General Hospital. *Arch Pathol Lab Med.* 2013;137(9):1237-46. http://dx.doi.org/10.5858/arpa.2012-0434-OA

9. Martin, B. R., Neiderer, K., & Dancho, J. F. Pilomatrixoma: An Infrequently Encountered Lesion on the Lower Extremity. *Journal of the American Podiatric Medical Association*. 2012, *102*(5), 417-418.

10. Chan JJ, Tey HL: Multiple pilomatricomas: case presentation and review of the literature . Dermatol Online J. 2010, 15:2