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Malignant pilar cyst in a young woman: Case report and literature review

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ABSTRACT

The patient was a young, 30-year-old woman presenting with a long standing painless scalp lesion with six month history of recent increase in size with associated pain. Clinically the swelling had benign features, therefore excisional biopsy was done. However histology revealed malignant pilar cyst.

Keywords: Malignant pilar cyst, Radiotherapy, Scalp, Wide local excision

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INTRODUCTION

Skin adnexal tumors (SAT), malignant adnexal tumors (MATS), or primary adnexal cutaneous tumors (PACS) are exceedingly rare neoplasms arising from cells of skin appendages like the hair or nails [1].

The cell of origin could be from either of the four adnexal cells or admixture of the cells. The cells arise from either the apocrine, eccrine, or pilosebaceous unit. The tumors arising from the skin appendages are large diverse and rare, and in particularly those of pilosebaceous units like the malignant pilar cyst are rarer.

The aim is to highlight the features of the tumor, the diagnostic dilemma, and treatment options as no standard treatment is outlined due to few documented cases in literature.

CASE REPORT

Our patient is a 30-year-old woman with a four year history of initially painless scalp swelling with recent rapid increase and pain. She had no symptoms in any of the systems, no medical diseases, no past surgeries.

The significant finding on physical examination was that of a well-circumcised scalp swelling about 8 cm by 4 cm, with regular edge, non-tender, attached to skin with no punctum but fluctuant, no cervical lymph nodes enlargement. An impression of a sebaceous cyst was made and the patient had an excisional biopsy via an elliptical incision over the cyst, which was dissected intact without rapture the specimen subjected to histology revealed:

1. Laminated keratin.
2. Presence of squamous epithelium lining exhibiting moderate to severe dysplasia.
3. Large cells with hyperchromatic mildly pleomorphic nuclei and prominent nucleoli.
4. Multiple areas of apparent invasion unto the wall of the cyst.
5. Presence of clusters and single cells within the cyst wall.
6. Mild chronic inflammatory cell.
7. Atypia confined to wall cyst.

Refer to Figures 1–5 for gross and pathology slides of the malignant pilar cyst.

Figure 1: Excised scalp cyst.

Figure 2: Unruptured cyst.

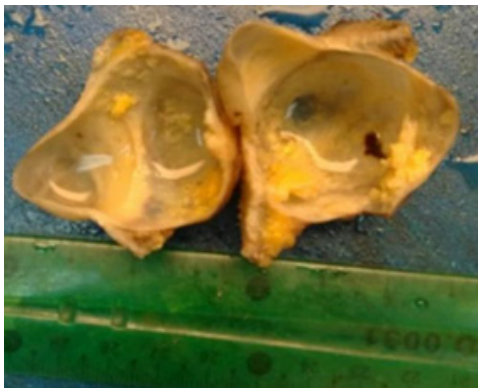

Figure 3: Bivalve cyst showing loculations.

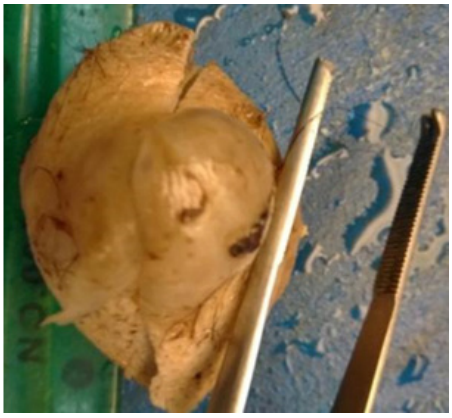

Figure 4: After opening cyst.

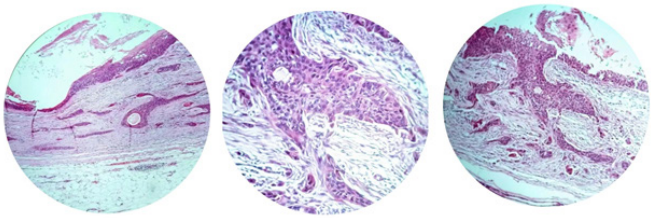

Figure 5: Histology slide of excised cyst wall showing laminated keratin, squamous epithelium with moderate to severe dysplasia, hyperchromasia of cells, pleomorphism of nucleus and prominent nucleolia. There is occasional invasion of cyst wall with single and clusters of such cells, accompanied by chronic inflammatory infiltrates with infrequent mitoses, however no invasion into the overlying epidermis which is unremarkable.

No involvement of overlying epidermis with the presence of malignant cells. The slides were referred for immunohistochemistry in a higher oncology center.

Results came positive for malignant pilar tumor.

The patient had computed tomography (CT) imaging of head and neck region which was unremarkable. She was advised for chemo and radiotherapy which she declined, had scar excision, no malignancy in scar. Follow-up for surveillance was scheduled after five years, no recurrence.

DISCUSSION

Cutaneous malignancies are the commonest human cancers, particularly in Caucasians [1–4]. However, the incidence varies geographically, due to the strong environmental association with these skin cancers, such as light, carcinogenic chemicals like arsenic, tobacco smoking, and genetic factors of white race [5–7].

Skin adnexal tumors (SAT) or malignant adnexal tumors (MAT) or primary adnexal cutaneous tumors (PAC) are exceedingly rare neoplasms. These tumors comprise only 1–2% of all skin malignancies [8].

Skin adnexal tumors have been linked with exposure to ultraviolet radiation and there are recognized syndromes like Muir–Torre syndrome, COWDEM, Brooke–Spiegler syndrome have been associated with these tumors.

The skin or integument is the largest human organ with surface area of 18 m^2 and constituting 15% of total body weight.

It serves the functions of protecting the body against ultraviolet light and microbes, maintains fluid and electrolyte balance, subserves metallic as well as immunologic and sensoneural mechanisms in human body.

Embryologically, skin begins to develop from the fifth to seventh weeks in utero. The epidermis is derived from the ectodermal layer while the dermis is derived from the mesoderm.

The skin adnexa, such as hair and nail, are from the ectoderm, histologically they comprise the pilosebaceous

and the apocrine and eccrine glands. The skin adnexal tumors, SAT, are from these skin appendages.

The hair is modified keratinized skin appendage, arising from the hair follicle, which is a tubular invagination from the epidermis. The hair shaft has an inner medulla, a middle cortex layer and outer cuticle.

The external root sheath is the outer most epidermal cell layers of hair bulb [9–12].

Malignant pilar tumors are exceedingly rare, with literature review citing only 50 published case reports [13]. Malignant pilar tumors (MAT) arise either de novo or in a pre-existing proliferating pilar cyst or pilar cyst [14, 15].

Acanthoma, trichophila melanoacanthoma, invasive hair matrix tumor, invasive pilomatrixoma, and giant hair matrix tumor are benign cystic lesions which typically occur on the scalp of elderly women [16]. They arise from outer root sheath of hair follicle [17]. Histologically, there is diffuse homogenous keratinization with round clear cells.

Proliferating trichilemmal cysts, from which majority of malignant pilar cysts arise, are usually lobulated cystic structures, comprise 30% of all pilar cysts [18, 19].

It is composed of mature keratinocytes lining keratin filled spaces. Among SAT, there are limited literature reviews on the follicular varieties like the malignant pilar cysts, which arise from the pilosebaceous unit, unlike the more common ones from the apocrine and eccrine sweat glands [20].

Literature review stated only about 50 cases reported previously, though rare, clinically they may be confused with the commoner–benign lesions such as pilar cysts or even sebaceous cyst and dismissed as such. However, vigilance is needed in their management as some can be aggressive and prone to locoregional and distant metastatic [21].

Malignant pilar cyst arises from the outer root sheath of the hair follicle, commonly seen in the elderly women between the fifth and sixth decade [16], mostly on the scalp [22].

Suspicious pilar cysts for malignancy are large size, nodulocystic lesions, irregular edges, ulcerative lesions, local lymphadenopathy, and extra-scalp lesions [23].

Also history of a rapid increase in size in a previously dormant lesion, as these lesions usually arise from existing long standing proliferating pilar cyst or de novo [13, 24].

Differential diagnoses include sebaceous cyst, the benign counterpart trichilemmal cyst, well differentiated squamous cell carcinoma can be confused with malignant pilar cyst, as well as most cutaneous lesions should be differentiated, especially those occurring on the scalp in elderly women.

Proper evaluation is necessary, though most of these lesions come to light only after biopsy with definitive histological results. Suspicious cutaneous lesions are subjected to relevant histological analysis after either excisional, trucut, wedge biopsy, or even fine needle aspiration cytology (FNAC) as demonstrated in one publication [25].

The proliferating pilar cyst has cystic spaces with trichilemmal keratinization, squamous cells lining, and peripheral palisading of the basal layer without granular layer. There may be occasional atypia with mitotic figures. This feature makes it difficult to differentiate from the malignant variety [26, 27].

The malignant pilar cyst on the other hand is characterized by the presence of irregularly arranged cells with or without lymphovascular or neural invasion infiltration, cells are horizontally oriented, with atypia, mitosis, and diminished sclerosing stroma and dyskeratosis, presence of palisading of cells, absence of epidermal lesion with eosinophilic malignant membrane. Presence of blank or ghost cells may be seen [28, 29].

Histologically attempts have been made by Ye et al. [30]. To categorize these tumors into three groups to serve as guide for treatment, predict possibility of local recurrence, and lymp-node metastasis as well as distant metastasis.

Group I

Unremarkable histology with no cellular atypia or ploidy or lymphovascular infiltration.

Recurrence at – 0%

Group II

Low grade malignancy

Irregular, locally invasive contours, with extension into the deep dermis. This group is locally aggressive. Recurrence is 15%

Group III

High grade malignancy with irregular contours, atypical mitotic figures, stromal desmoplasia with absence of lobular pattern, necrosis may be present. Recurrence up to 50%.

Histologically malignant pilar cyst may be indistinguishable from squamous cell carcinoma (MPC) usually display trichilemmal keratinization with palisading, presence of hyaline basement membrane, and if arising from a pre-existing pilar cyst thus can be demonstrated [31].

Immunohistochemistry may be employed in difficult circumstances, to exclude other differential diagnosis though there is a vast array of immunohistochemistry makers, for cutaneous malignancies, they are generally non-specific and there is a lot of overlap for positivity amongst these tumors.

Non-specific tumor makers reported to be expressed by the malignant tumors included P53, K1–67 [32–35].

Antibodies to certain keratins selective for follicular epithelium such as AE3 and AE14 may assist in differentiating the well differentiated squamous cell

carcinoma from the malignant pilar tumors, since histology may be confusing [34, 36].

Regardless, if there is a high index of suspicion, based on clinicopathological features, the best management approach is a multidisciplinary one, constituting the relevant surgical specialty as well as, oncologist, dermatologist, radiologist, and members of tumor board where available.

Other researches have proposed use of similar agents and regiment for breast cancer for apocrine tumors, as they save similar cell of origin with the breast.

Treatment

Treatment of these rare lesions is basically similar to skin malignancies and in general to all oncological diseases. A multidisciplinary approach involving the relevant surgical specialties like the ear, nose, throat (ENT), General Surgeons, Dermatologist, Oncologist, Tumor Board, Pathologist, and many more, may be required to participate in the design of the plan.

However surgery is unanimously employed as the mainstay [36, 37]. Wide local excision is done and margins of up to 2 cm suggested [37, 38]. Mohr's micrographic surgery is offered in certain situations to cosmetically sensitive areas [39–42].

Radical surgeries like amputations for limb lesions have been suggested to avoid distant metastasis to chest and even brain [43, 44]. The role of sentinel lymph node biopsy is not yet well established [45].

Less invasive procedures like use of laser, cryosurgery, retinoic acid, and topical chemotherapeutic agents have been suggested [46, 47].

Radiotherapy has been widely used and offers some ensuring outcomes in the treatment of skin adnexal tumors as an adjunct [45, 48].

There is a lot of uncertainty in many instances where chemotherapy was used as the results were inconclusive, in terms of response, recurrence free, and increased diseases free survival rates [49].

Some researchers proposed the use of similar chemotherapy used for breast cancer for treatment of apocrine malignant neoplasms due to similarity of cell of origin [37, 50].

CONCLUSION

The watchword for the management of these rare malignant adnexal tumors is to have high index of suspicion, though rare, misdiagnosis can lead to morbidity and mortality as some are aggressive and in few cases recurrence has been reported after many years. Hence, the need for vigorous follow-up and surveillance, this will also help in enriching our knowledge in the best possible way to cater for these patients.

REFERENCES

1. Lomas A, Leonardi-Bee J, Bath-Hextall F. A systematic review of worldwide incidence of nonmelanoma skin cancer. Br J Dermatol 2012;166(5):1069–80.
2. Almahroos M, Kurban AK. Ultraviolet carcinogenesis in nonmelanoma skin cancer. Part I: Incidence rates in relation to geographic locations and in migrant populations. Skinmed 2004;3(1):29–35.
3. Stern RS. Prevalence of a history of skin cancer in 2007: Results of an incidence-based model. Arch Dermatol 2010;146(3):279–82.
4. Rogers HW, Weinstock MA, Feldman SR, Coldiron BM. Incidence estimate of nonmelanoma skin cancer (keratinocyte carcinomas) in the U.S. population, 2012. JAMA Dermatol 2015;151(10):1081–6.
5. Leiter U, Garbe C. Epidemiology of melanoma and nonmelanoma skin cancer—The role of sunlight. Adv Exp Med Biol 2008;624:89–103.
6. Rigel DS. Cutaneous ultraviolet exposure and its relationship to the development of skin cancer. J Am Acad Dermatol 2008;58(5 Suppl 2):S129–32.
7. Jones RR. Ozone depletion and its effects on human populations. Br J Dermatol 1992;127 Suppl 41:2–6.
8. Stam H, Lohuis PJFM, Zupan-Kajcovski B, Wouters MWJM, van der Hage JA, Visser O. Increasing incidence and survival of a rare skin cancer in the Netherlands. A population-based study of 2,220 cases of skin adnexal carcinoma. J Surg Oncol 2013;107(8):822–7.
9. Sato K, Kang WH, Saga K, Sato KT. Biology of sweat glands and their disorders. I. Normal sweat gland function. J Am Acad Dermatol 1989;20(4):537–63.
10. Elder DE, Elenitsas R, Johnson BL, Murphy GF. Lever's Histopathology of the Skin. 9ed. Baltimore (MD): Lippincott Williams & Wilkins; 2005.
11. Yousef H, Miao JH, Alhajj M, Badri T. Histology, Skin Appendages. In: StatPearls. Treasure Island (FL): StatPearls Publishing; 2024.
12. Brown TM, Krishnamurthy K. Histology, Hair and Follicle. In: StatPearls. Treasure Island (FL): StatPearls Publishing; 2024.
13. Garg PK, Dangi A, Khurana N, Hadke NS. Malignant proliferating trichilemmal cyst: A case report with review of literature. Malays J Pathol 2009;31(1):71–6.
14. Saida T, Oohara K, Hori Y, Tsuchiya S. Development of a malignant proliferating trichilemmal cyst in a patient with multiple trichilemmal cysts. Dermatologica 1983;166(4):203–8.
15. Takata M, Rehman I, Rees JL. A trichilemmal carcinoma arising from a proliferating trichilemmal cyst: The loss of the wild-type p53 is a critical event in malignant transformation. Hum Pathol 1998;29(2):193–5.
16. Kemaloğlu CA, Öztürk M, Aydın B, Canöz Ö, Eğilmez O. Malignant proliferating trichilemmal tumor of the scalp: Report of 4 cases and a short review of the literature. Case Reports Plast Surg Hand Surg 2022;9(1):158–64.
17. Headington JT. Tumors of the hair follicle. A review. Am J Pathol 1976;85(2):479–514.
18. Jones EW. Proliferating epidermoid cysts. Arch Dermatol 1966;94(1):11–9.

19. Satyaprakash AK, Sheehan DJ, Sangüeza OP. Proliferating trichilemmal tumors: A review of the literature. Dermatol Surg 2007;33(9):1102–8.
20. Klein W, Chan E, Seykora JT. Tumors of the epidermal appendages. In: Elder DE, editor. Lever's Histopathology of the Skin. 9ed. Philadelphia, PA: Lippincott Williams & Wilikins; 2005. p. 867–926.
21. Hiramatsu K, Sasaki K, Matsuda M, et al. A case of trichilemmal carcinoma with distant metastases in a kidney transplantation patient. Transplant Proc 2015;47(1):155–7.
22. Durairaj AR, Mahipathy SRRV, Vivakaran TTR, Harikrishnan V, Esakki M. Malignant proliferative trichilemmal tumour of the nape of the neck – A case report. J Clin Diagn Res 2016;10(2):PD19–20.
23. Rao S, Ramakrishnan R, Kamakshi D, Chakravarthi S, Sundaram S, Prathiba D. Malignant proliferating trichilemmal tumour presenting early in life: An uncommon feature. J Cutan Aesthet Surg 2011;4(1):51–5.
24. Kini JR, Kini H. Fine-needle aspiration cytology in the diagnosis of malignant proliferating trichilemmal tumor: Report of a case and review of the literature. Diagn Cytopathol 2009;37(10):744–7.
25. Ramaswamy AS, Manjunatha HK, Sunilkumar B, Arunkumar SP. Morphological spectrum of pilar cysts. N Am J Med Sci 2013;5(2):124–8.
26. Ackerman AB, Viragh PA, Chongchitnant N. Proliferating follicular cystic neoplasm. In: Ackerman AB, Viragh PA, Chongchitnant N, editors. Neoplasms with Follicular Differentiation. Philadelphia: Lea & Febiger; 1993. p. 553–70.
27. Lund HZ. Atlas of Tumour Pathology. Tumours of the Skin. Washington: Armed Forces Institute of Pathology; 1957.
28. Noto G. 'Benign' proliferating trichilemmal tumour: Does it really exist? Histopathology 1999;35(4):386–7.
29. Sakamoto F, Ito M, Nakamura A, Sato Y. Proliferating trichilemmal cyst with apocrine-acrosyringeal and sebaceous differentiation. J Cutan Pathol 1991;18(2):137–41.
30. Ye J, Nappi O, Swanson PE, Patterson JW, Wick MR. Proliferating pilar tumors: A clinicopathologic study of 76 cases with a proposal for definition of benign and malignant variants. Am J Clin Pathol 2004;122(4):566–74.
31. Agarwal C, Pujani M, Raychaudhuri S, Arora S, Rana D, Chauhan V. Squamous cell carcinoma versus malignant proliferating trichilemmal tumor: A histopathological dilemma with review of literature. Indian J Dermatol 2019;64(4):339.
32. Alhumaidi A. Practical immunohistochemistry of epithelial skin tumor. Indian J Dermatol Venereol Leprol 2012;78(6):698–708.
33. Alici O, Keles MK, Kurt A. A rare cutaneous adnexal tumor: Malignant proliferating trichilemmal tumor. Case Rep Med 2015;2015:742920.
34. Kandasamy S, Murugan R, Vasugi GA, Gunasekaran KP. Histomorphological patterns of skin adnexal tumors with an insight into molecular updates – A single institutional study. J Pathol Nep 2023;13(1):2008–12.
35. LeBoit PR, Burg G, Weedon D, Sarasain A, editors. World Health Organization Classification of Tumours. Pathology and Genetics of Skin Tumors. Lyon: IARC Press; 2006.
36. Kibbi N, Owen JL, Worley B, et al. Evidence-based clinical practice guidelines for extramammary paget disease. JAMA Oncol 2022;8(4):618–28.
37. Sau P, Graham JH, Helwig EB. Proliferating epithelial cysts. Clinicopathological analysis of 96 cases. J Cutan Pathol 1995;22(5):394–406.
38. Morgado B, Agostini P, Rivero A, Silva N. Extensive and ulcerated malignant proliferating trichilemmal (pilar) tumour, arising from multiple, large, degenerated trichilemmal (pilar) cysts. BMJ Case Rep 2016;2016:bcr2015209785.
39. Tierney E, Ochoa MT, Rudkin G, Soriano TT. Mohs' micrographic surgery of a proliferating trichilemmal tumor in a young black man. Dermatol Surg 2005;31(3):359–63.
40. Hamman MS, Brian Jiang SI. Management of trichilemmal carcinoma: An update and comprehensive review of the literature. Dermatol Surg 2014;40(7):711–7.
41. Fieleke DR, Goldstein GD. Malignant proliferating trichilemmal tumor treated with Mohs surgery: Proposed protocol for diagnostic work-up and treatment. Dermatol Surg 2015;41(2):292–4.
42. Lobo L, Amonkar AD, Dontamsetty VVSMK. Malignant proliferating trichilemmal tumour of the scalp with intra-cranial extension and lung metastasis—A case report. Indian J Surg 2016;78(6):493–5.
43. Park BS, Yang SG, Cho KH. Malignant proliferating trichilemmal tumor showing distant metastases. Am J Dermatopathol 1997;19(5):536–9.
44. Yi HS, Sym SJ, Park J, et al. Recurrent and metastatic trichilemmal carcinoma of the skin over the thigh: A case report. Cancer Res Treat 2010;42(3):176–9.
45. Cecchi R, Rapicano V, De Gaudio C. Malignant proliferating trichilemmal tumour of the scalp managed with micrographic surgery and sentinel lymph node biopsy. J Eur Acad Dermatol Venereol 2008;22(10):1258–9.
46. Jo JH, Ko HC, Jang HS, Kim MB, Oh CK, Kwon KS. Infiltrative trichilemmal carcinoma treated with 5% imiquimod cream. Dermatol Surg 2005;31(8 Pt 1):973–6.
47. Nakai N, Takenaka H, Hamada S, Kishimoto S. Identical p53 gene mutation in malignant proliferating trichilemmal tumour of the scalp and small cell carcinoma of the common bile duct: The necessity for therapeutic caution? Br J Dermatol 2008;159(2):482–5.
48. Amaral AL, Nascimento AG, Goellner JR. Proliferating pilar (trichilemmal) cyst. Report of two cases, one with carcinomatous transformation and one with distant metastases. Arch Pathol Lab Med 1984;108(10):808–10.
49. Siddha M, Budrukkar A, Shet T, et al. Malignant pilar tumor of the scalp: A case report and review of literature. J Cancer Res Ther 2007;3(4):240–3.
50. Battistella M, Mateus C, Lassau N, et al. Sunitinib efficacy in the treatment of metastatic skin adnexal carcinomas: Report of two patients with hidradenocarcinoma and trichoblastic carcinoma. J Eur Acad Dermatol Venereol 2010;24(2):199–203.

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Author Contributions

Ali Ibrahim Ali Hegy – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Amina Ibrahim El-yakub – Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Yaser Taha Sidahmed – Conception of the work, Design of the work, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Guarantor of Submission

The corresponding author is the guarantor of submission.

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Written informed consent was obtained from the patient for publication of this article.

Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.