Malignant proliferating trichilemmal tumor: a case report of a rare clinicopathological conundrum

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Received: 15 June 2021
Accepted: 3 December 2021

Proliferating trichilemmal tumors are rare, generally solitary, benign tumors arising from the hair's outer root sheath with trichilemmal keratinization. These tumors are predominantly found on the head, neck, and face of elderly females; the inguinal region is a highly unusual site of involvement. Malignant change in this tumor is rare, histologically mimicking squamous cell carcinoma. Metastatic malignant proliferating trichilemmal tumor requires a high index of suspicion for diagnosis and accurate histopathological reporting for timely intervention. Here, we report the case of a 65-year-old man presenting with nodular-ulcerative lesions in the groin, clinically confused with hidradenitis suppurativa and histologically with squamous cell carcinoma. He was finally diagnosed with an inoperable metastatic stage of malignant proliferating trichilemmal tumor and was treated with palliative care. This case highlights the innocuous clinical appearance of such lesions that might delay the management of this fulminant tumor.

Keywords: neoplasms, malignant neoplasm, tumor

Iran J Dermatol 2023; 26: 85-89

DOI: 10.22034/ijd.2021.290804.1385

INTRODUCTION

Outer root sheath tumors are uncommon appendageal tumors of hair origin showing squamoid cytological features with a trichilemmal keratinization pattern [1]. While trichilemmal cysts are often encountered, a proliferating trichilemmal tumor (PTT) is an uncommon, benign type with rare malignant transformation [2]. The most frequent sites are the scalp, neck, and face, with occasional involvement of the trunk, back, buttocks, and vulva [3]. Clinical similarity to various innocuous disorders and histological resemblance to squamous cell carcinoma poses diagnostic and therapeutic difficulties, contributing to delayed diagnosis [1,4]. Here, we report the case of a 65-year-old man presenting with nodular-ulcerative lesions in the groin, clinically confused with hidradenitis suppurativa and histologically with squamous cell carcinoma. The lesions were later diagnosed as advanced, metastatic stage malignant proliferating trichilemmal tumor

Please cite this article as: Chavan RB, Belgaumkar VA, Deshmukh N, Nikam B, Joshi VD. Malignant proliferating trichilemmal tumor: a case report of a rare clinicopathological conundrum. Iran J Dermatol. 2023;26(2):85-89. doi: 10.22034/ijd.2021.290804.1385.

(MPTT) arising at a rare site.

CASE PRESENTATION

A 65-year-old male presented to our tertiary care hospital with multiple swellings and foul-smelling ulcers over the inguinal region, commencing two months earlier. He had noticed three to four almond-sized swellings in his groin six months ago, rapidly increasing in size and number with ulceration and significant weight and appetite loss. On examination, he was extremely cachectic and pale with bilaterally enlarged firm, non-tender inguinal lymphadenopathy. Multiple firm nodules with crateriform ulceration were seen over bilateral groins and inner thighs with purulent discharge (Figure 1). There were no lesions elsewhere. Systemic examination was unremarkable except for mild hepatomegaly.

Scrutiny of his past records disclosed that his initial lesions had been treated in a private hospital as hidradenitis suppurativa based on clinical examination and histopathological changes of acute suppurative granulomatous inflammation. The lesions progressed despite multiple courses of intravenous antibiotics, and eventually, he was referred to us.

We performed biopsies from the nodule and ulcer edge, considering the differential diagnoses of hidradenitis suppurativa, scrofuloderma, and actinomycosis, prompted by the clinical appearance of discharging sinuses in the inguinal region. Histopathological findings suggested well-differentiated squamous cell carcinoma (Figure 2).

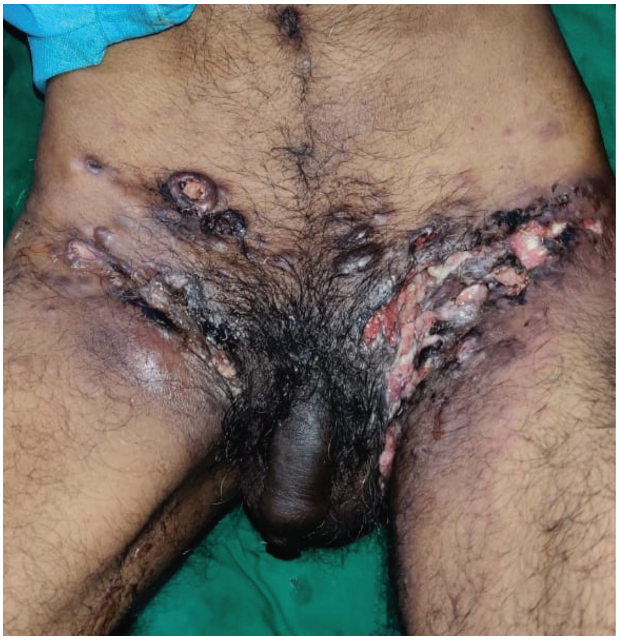

Figure 1. Multiple firm nodules with crateriform deep ulcerations and a purulent, foul-smelling discharge in the inguinal region.

Contrast-enhanced computed tomography (CECT) scan (abdomen, pelvis, and thorax) revealed multiple heterogeneously enhancing opacities in lymph nodes

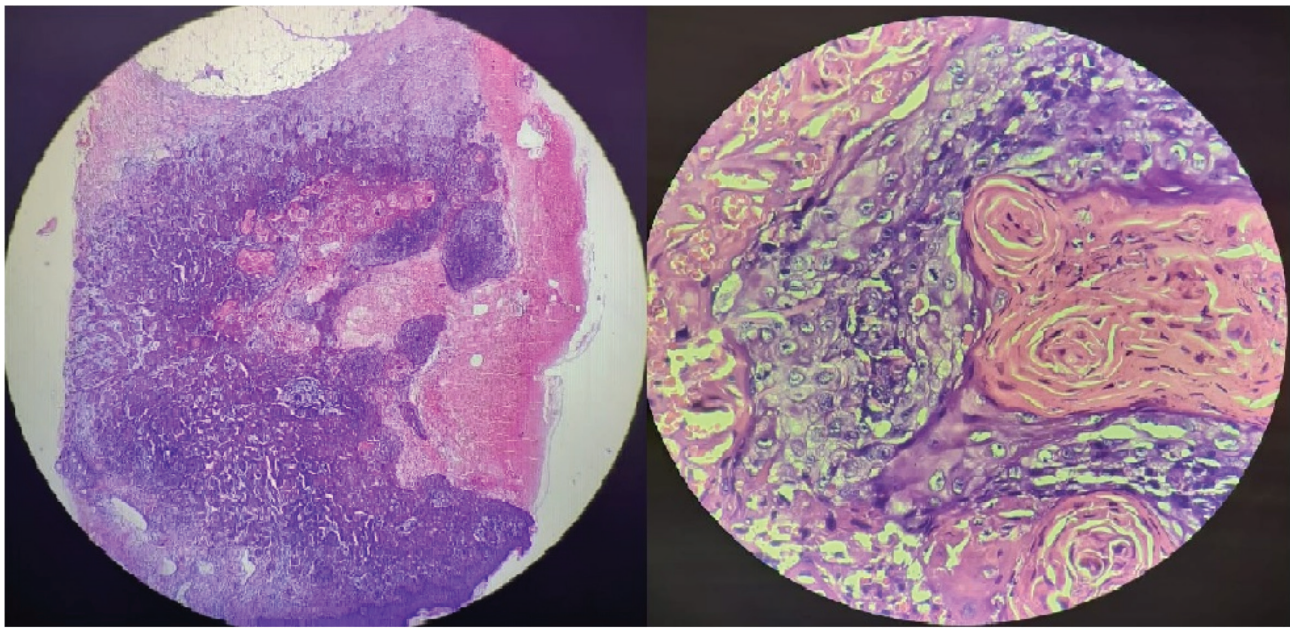

Figure 2. Hematoxylin and eosin-stained photomicrographs: infundibular proliferation (×4) and trichilemmal keratinization (×40)

and soft tissues of bilateral inguinal regions, penis, and scrotum (Figure 3), bilateral lung fields, and thyroid gland, with mediastinal lymphadenopathy and thrombosis of the common femoral vein and iliac veins extending into the superficial femoral vein, suggestive of metastasis.

Due to the baffling clinicopathological discrepancy, the clinical images of his initial lesions were retrieved, showing skin-colored nodules in the lower abdominal and inguinal region resembling sinuses (Figure 4). Serial biopsy sections from these lesions (at first presentation) showed acute suppurative granulomatous inflammation (Figure 5). The histopathological review of the current biopsy specimens (Figure 2) showed characteristic features of malignant proliferating trichilemmal tumor: infundibular proliferation, nests of atypical squamoid cells with increased mitoses, peripheral palisading, and invasive borders showing abrupt trichilemmal keratinization (mimicking keratin pearls) without stratum granulosum. The immunohistochemistry (IHC) markers Ki-67 and CD 34 (specific for the outer hair root sheath) wer, e positive.

Thus, a final diagnosis of an inoperable malignant proliferating trichilemmal tumor with extensive metastases was made. The patient was referred to a palliative care center for further management.

Ethical consideration

Written informed consent was obtained from patient for publication, Ethics code: Not applicable.

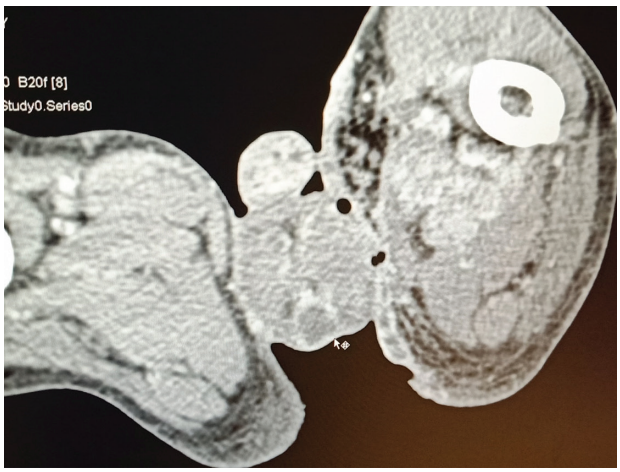

Figure 3. Contrast-enhanced computed tomography scan image of the pelvis showing multiple heterogenous necrotic hypodense foci in the scrotum (white cursor arrow) and soft tissues.

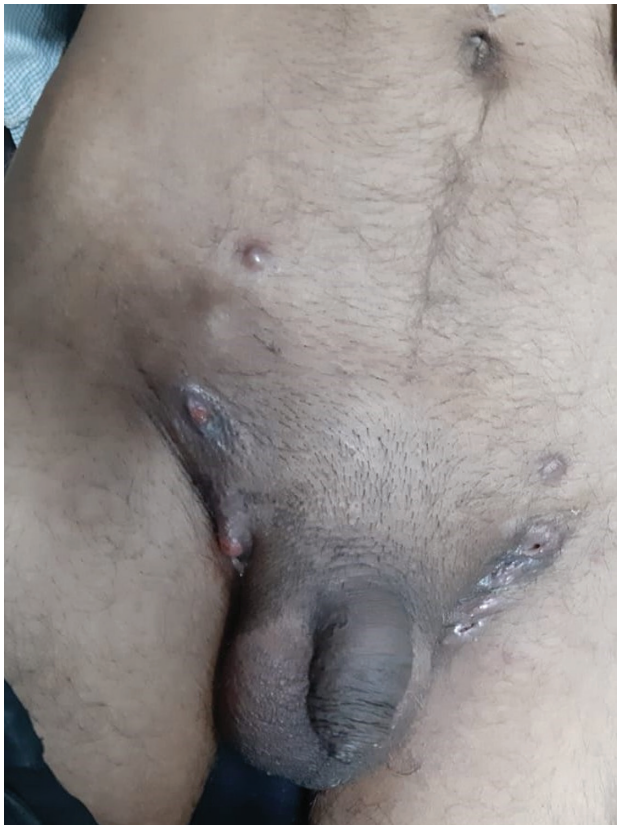

Figure 4. Initial clinical presentation [image captured two months before Figure 1] with few skin-colored nodules over the inguinal and lower abdominal region resembling sinuses.

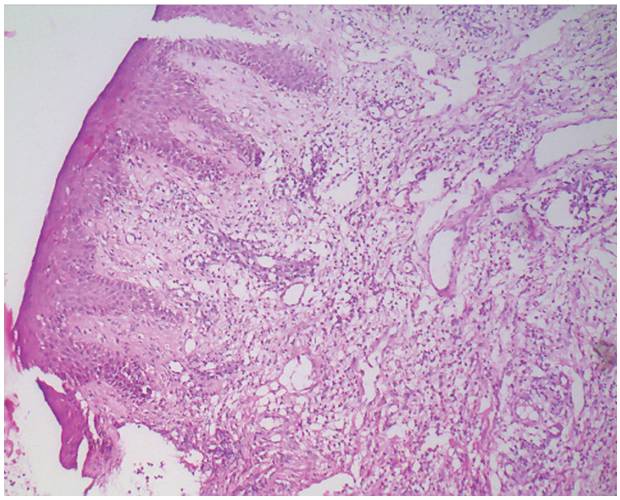

Figure 5. Low-power photomicrograph of biopsy from initial lesions (skin-colored nodules) showing non-specific changes of acute suppurative granulomatous inflammation.

DISCUSSION

First described by Wilson Jones in 1966, proliferating trichilemmal tumor (PTT, *syn. proliferating pilar cyst, giant hair matrix tumor* [2])

is a rare, generally solitary, benign, solid or cystic appendageal tumor with a predilection for females in the fourth to eighth decades of life [1]. It originates from the outer root sheath of the hair follicle, either de novo or from a pre-existing pilar cyst wall [2]. Though the preferred sites are the scalp, neck, and face, it may also be seen over the trunk, back, and buttocks [2,3]. In its solid form, it presents as single or rarely multiple nodules with indolent growth over months to years, forming a lobulated or exophytic mass that occasionally ulcerates [1]. The cystic form clinically mimics a sebaceous or keratinous cyst. Histopathologically, it shows dermal sheets, strands and lobules of hyperplastic squamous epithelium with peripheral palisading, squamoid cells without stratum granulosum, and characteristic abrupt trichilemmal keratinization [4]. Trauma and inflammation may convert a trichilemmal cyst into a proliferating variant [2]. Increased mitoses, aneuploidy, cellular atypia, angiolymphatic and perineural invasion, and stromal infiltration are microscopic signs of malignant transformation [5-7].

A malignant proliferating trichilemmal tumor (MPTT) bears close histological resemblance to squamous cell carcinoma (SCC), from which it can be distinguished by immunohistochemistry markers like AE-13, AE-14, calretinin, and CD-34 [8]. MPTT is known for its pronounced tendency for tissue invasion and distant metastases compared to its histopathological mimicker, squamous cell carcinoma. Therapeutic options include wide surgical excision with or without radiotherapy and chemotherapy, with some reports depicting the use of platinum compounds, vinca alkaloids, and IFN-alpha [9].

In our patient, the nodules originated in the groin—an extremely rare site with only a few documented cases [10]. The initial presentation with ulcerated nodules and exophytic sinuses was clinically confounding and led to the erroneous differential diagnoses of hidradenitis suppurativa, actinomycosis, and scrofuloderma. To further compound the dilemma, the first biopsy showed non-specific acute suppurative granulomatous inflammation (attributable to secondary change or a non-representative biopsy site), while the repeat biopsy was reported as SCC. This discordance (the clinical features were inconsistent with SCC) and the detection of extensive metastasis induced us to revisit the diagnosis and perform a meticulous clinicopathological review, establishing the diagnosis of a malignant proliferating trichilemmal tumor. Unfortunately, the inadvertent diagnostic delay and fulminant course precluded all therapeutic options except palliative care.

It is worth speculating whether wide surgical excision of the initial benign lesions might have averted this catastrophic evolution. Ominous risk factors for aggressive biologic behavior of PTT are non-scalp site, multiplicity, size more than 5 cm, atypia, and mitotic activity, all of which were present in our case [1,2]. One of the two cases of inguinal proliferating pilar cysts reported by Amaral *et al.* showed transformation to epidermoid carcinoma with a fatal outcome due to generalized metastases despite surgical resection [10]. The authors proposed that a proliferating pilar cyst should be diagnosed in pilar cysts with epithelial proliferation showing well-defined borders and abundant keratinization despite cytologic atypia and mitotic activity. On the other hand, diagnosing carcinoma in proliferating pilar cysts should be reserved for lesions showing poorly defined borders and clear-cut infiltrative properties.

CONCLUSION

Malignant proliferating trichilemmal tumor, a rare, highly invasive adnexal tumor, poses a clinicopathological conundrum, particularly when presenting with atypical morphology at unusual sites. The present case emphasizes the importance of a high index of suspicion supported by vigilant and accurate histopathological interpretation of multiple tissue specimens from various involved sites to facilitate early diagnosis and timely intervention.

Acknowledgments

The authors gratefully acknowledge our institute's Departments of Pathology and Radiodiagnosis for their valuable contribution to evaluating and diagnosing the patient.

Authors contributions

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Management of case : Vasudha Belgaumkar, Nitika Deshmukh, Vijay Joshi

Drafting of manuscript : Vijay Joshi, Vasudha Belgaumkar

Critical revision of the manuscript for important intellectual content: Vasudha Belgaumkar, Balakrishna Nikam, Nitika Deshmukh, Vijay Joshi

Funding source

None.

Conflict of interest: None declared.

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