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CASE REPORT

Malignant proliferating trichilemmal cyst: a case report with review of literature

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Abstract

Proliferating trichilemmal cyst is a rapidly growing large cutaneous adnexal neoplasm occurring on the head and neck region of elderly women. Malignant transformation has rarely been reported in these lesions. We describe here a 85-year-old lady who presented with a large ulcerated growth over the scalp for one year duration. Incisional biopsy revealed proliferating trichilemmal cyst with malignant transformation. She underwent wide local excision of this growth. She is alive and without evidence of disease after 14 months of follow up. Because of limited number of cases reported in literature, management of malignant proliferating trichilemmal cyst is controversial. Treatment mainly entails wide local surgical excision. Many other adjuvant modalities have been tried. This paper presents the diagnosis and management of one case of malignant proliferating trichilemmal cyst followed by review of the literature.

Keywords: proliferating trichilemmal cyst, cutaneous adnexal neoplasm, malignant trichilemmal cyst

INTRODUCTION

Proliferating trichilemmal cyst is a rare, usually benign, tumour of external root sheath derivation and in most cases appears to develop within the wall of a pre-existenting pilar cyst. Proliferating trichilemmal cyst is more common in elderly women than men and is usually localized to the scalp. The tumour shows trichilemmal keratinisation without interposed granular layer. If there is frank invasion into adjacent structures in association with anaplasia and tumour necrosis, the diagnosis of malignant proliferating trichilemmal cyst is appropriate. Although rare, malignant transformation in a proliferating trichilemmal cyst is a distinct entity. Therefore every cystic mass of the scalp should be excised and must be subjected to histopathological examination.

CASE HISTORY

An 85-year-old lady presented to our hospital with a large ulcerated growth of the scalp of 1 year duration, progressively increasing in size (FIG. 1). The patient was otherwise healthy with no significant past medical history. No history suggestive of trauma and chronic irritation except hair combing was present. On examination there was a single, ulcerated scalp tumour of 15 × 8 cm in diameter at the right parieto-occipital region. It was not fixed to the underlying bone but there was one tender lymph node of 1.5 cm size palpable in the right suboccipital region. Chest roentgenogram did not reveal any evidence of pulmonary metastasis. Contrast-enhanced CT scans of the brain also did not show any evidence of intracranial invasion. An incisional biopsy done was suggestive of malignant proliferating trichilemmal cyst. Fine needle aspiration cytology of the enlarged lymph node revealed acute inflammatory changes. Surgical excision of the tumour with 2 centimetres margin along with lymph node excision was performed.

Pathology:

Histological evaluation of the excised specimen was consistent with the diagnosis of malignant proliferating trichilemmal cyst. Grossly, the

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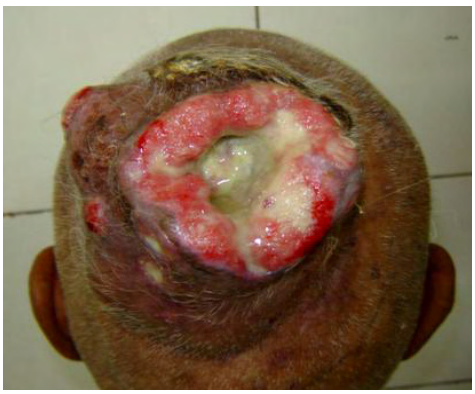

FIG. 1: Ulcerated malignant proliferating trichilemmal cyst of the scalp.

tumour was nodular, circumscribed, and protruding through the ulcerated hairy skin. Microscopically, the mid dermis, deeper dermis and subcutaneous tissue showed cellular tumour comprised of lobules of squamous cells with centres of lobules filled with keratinous material derived from abrupt keratinisation of large polygonal cells with abundant pale eosinophilic cytoplasm (trichilemmal type). The cells showed moderate to marked pleomorphism, high mitotic activity with abnormal mitotic figures, tumour giant cells and foci of invasion into surrounding tissue (FIG. 2). All the resected margins were free of tumour. The lymph node showed only reactive changes. A diagnosis of malignant proliferating trichilemmal cyst was made. At 14 months of follow up, there was no evidence of local recurrence or distant metastasis.

DISCUSSION

Cutaneous tumours derived from the outer root sheath of hair follicles, which show trichilemmal keratinisation, are trichilemmal cysts, proliferating trichilemmal cysts and malignant proliferating trichilemmal cysts. A trichilemmal cyst is by far the most common of these. These cysts are mainly seen on the scalp and are relatively common. They well circumscribed in the dermis with stratified peripheral cells, which rest on thickened basement membrane. Centrally, the cells enlarge

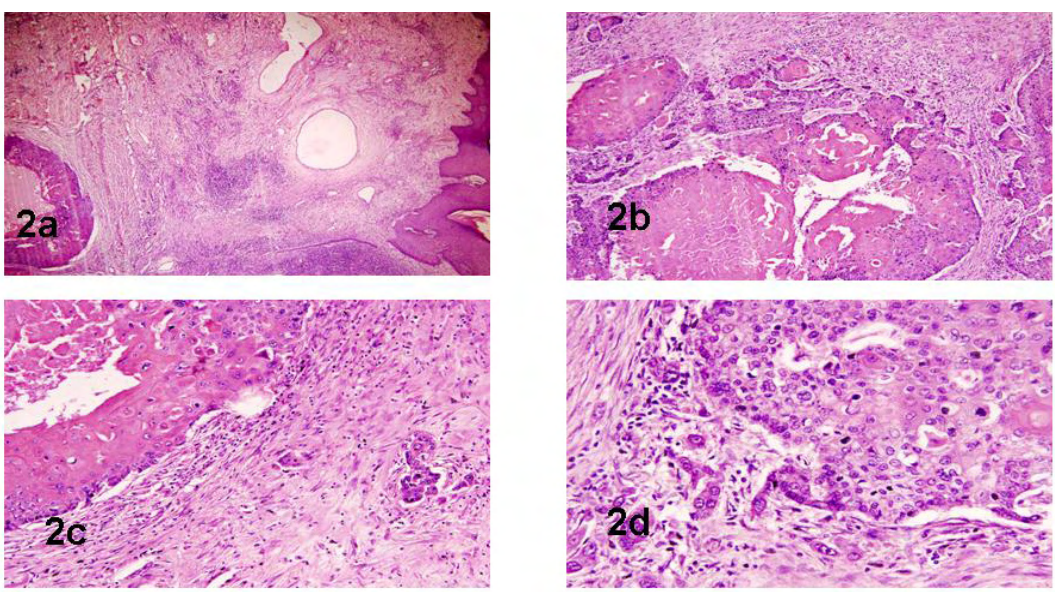

FIG. 2: (a) Tumour islands seen in the deep dermis. Upper dermis and epidermis are uninvolved (H&E x40). (b) Large deposits of keratinous material surrounded by pleomorphic squamous cells (H&E x 250). (c) Trichilemmal keratinisation without interposed granular layer. (H&E x 250). (d) Cells show anaplasia, high N:C ratio, mitosis and invasion into the connective tissue (H&E x400).

and accumulate abundant intracellular glycogen, with a resultant pale glossy appearance. There is then formation of a dense keratin layer without interposition of a granular cell layer known as trichilemmal keratinisation.[1,2]

Proliferating trichilemmal cyst is a rare but morphologically distinctive tumour usually occuring in the scalp of elderly women. Proliferating trichilemmal cyst has been reported under a variety of names including giant hair matrix tumour, invasive pilomatrixoma, proliferating epidermoid cyst, pilar tumour of the scalp, trichilemmal pilar tumour, trichochlamydocarcinoma, proliferating trichilemmal cyst, proliferating trichilemmal tumour, and proliferating follicular cystic neoplasm. Browstein *et al*[2] presented a series of 50 cases of proliferating trichilemmal cysts. They described proliferating trichilemmal cysts microscopically, as well-circumscribed growths in the corium containing anastomosing sheets and strands of hyperplastic epithelium, forming solid areas and small and large cysts. They concluded that the most characteristic feature was trichilemmal keratinisation identical to that seen in ordinary trichilemmal cysts. Peripheral cells were palisaded and rested on a thick, hyalinized basement membrane, did not flatten and form a granular layer before keratinising, but enlarged, became pale staining and glycogen rich and abruptly developed into dense horn. They observed a spectrum from trichilemmal cyst with minimal hyperplasia to full-blown proliferating trichilemmal cyst. The similarities between trichilemmal cyst and proliferating trichilemmal cyst, identification of intermediate stages between the two, and the occasional association of both the conditions suggest that proliferating trichilemmal cyst is a complication of trichilemmal cyst. It would seem most likely that trauma and inflammation induce an occasional trichilemmal cyst to proliferate in a less stereotyped manner, showing a much broader range of pilosebaceous differentiation than the ordinary trichilemmal cyst, as well as a wider degree of cellular atypia, yet maintaining its benign biological behaviour. However, proliferating trichilemmal cyst can occur *de novo* without a pre-existing lesion.

Malignant proliferating trichilemmal cyst is the rarest of trichilemmal tumours. Headington[3] proposed the term "malignant proliferating trichilemmal cyst" for the proliferating trichilemmal cyst with malignant transformation. The malignant proliferating trichilemmal cyst most often arises in a proliferating trichilemmal cyst, a benign tumour that is the main consideration in the differential diagnosis of malignant proliferating trichilemmal cyst. The distinguishing feature of the malignant tumour is the addition of a frankly invasive component that may retain a clear cell pattern with classical trichilemmal keratinisation. Nuclear and mitotic activities are variable. Vascular and/or perineural invasion may be observed. The real incidence of a malignant proliferating trichilemmal cyst is unknown, due to its rarity and also to inconsistencies in nomenclature and misclassification as squamous cell carcinoma. Herrero *et al*[4] demonstrated loss of CD34 immunohistochemical reactivity (CD34 considered to be a marker of outer root sheath differentiation and has been shown to be present in proliferating trichilemmal cyst) and concluded that absence of CD34 immunohistochemical reactivity should be considered to be a consequence of the undifferentiation of most tumour cells.

Kim *et al*[5] showed malignant proliferating trichilemmal tumours can manifest as either a cystic or solid mass on imaging studies. When solid, signal intensities of the tumour on MR images were grossly the same as those of most other soft-tissue tumours: areas of hypointensity on T1-weighted images, hyperintensity on T2-weighted images, and substantial enhancement after contrast. Poorly defined margins as well as penetration of the tissue planes suggest the malignant nature of the tumour. The tumour is typically benign-looking on CT scans if it is cystic. Malignant radiographical characteristics include local invasion into the calvaria, meninges, and dural sinuses. Other findings suggestive of malignancy on imaging may include enhancement of the soft-tissue components in the periphery of the cyst and infiltration of the surrounding soft tissues. CT is the technique of choice to monitor bony erosion, whereas MR imaging would be more suitable to assess soft-tissue infiltration and dural involvement.[6]

Smooth soft-tissue elevations from the inner wall of the mass are an important clue in predicting the nature of the mass, not a pure cyst but a cystic tumour. These areas corresponded histologically to proliferating lobules of epithelium, characteristic of malignant proliferating trichilemmal tumour. Jung *et al*[7] suggested the role of 18F-fluorodeoxyglucose-positron emission tomography in malignant proliferating trichilemmal tumour cases to

TABLE 1. Published reports of malignant proliferating trichilemmal cyst

Study	No. of cases	Sex	Age		Size (cm)	Duration (Years)	Treatment given	Outcome	Follow-up (mo)
Kim HJ et al (5)	1	M	75	Lip	2.7	4	Excision	No recurrence	NS
Jung et al (7)	1	F	69	Scalp	2	1	Mohs micrographic surgery	Local recurrence	2
Alvarez et al (9)	1	F	84	Scalp	3.5	15	Excision	No recurrence	24
Arico et al (10,11)	1	F	42	Scalp	2	1	Excision + RT+ CT	Lymph node metastases DOD	161
	2	F	75	Groin	2	Unknown	Excision + RT+ CT	Lymph node metastases DOD	12
		F	52	Groin	8	2	Excision + RT+ CT	Lymph node metastases DOD	1
Batman et al (12)	1	F	71	Scalp	1.8	NS	Excision + RT	Soft tissue metastases;	97
Fernando et al (13)	1	F	97	Scalp	3	20	Excision	No recurrence	6
Herrero et al (4)	3	F	64	Scalp	3	NS	NS	NS	19
		M	53	Scalp	3	NS	NS	NS	18
		F	66	Scalp	2.5	NS	NS	NS	NA
Hayashi et al (14)	1	M	50	Neck	3	3	CT	DOD	4
Jaworski (15,16)	1	F	87	Scalp	NS	1	Excision	No recurrence	NS
Mehregan et al (17)	3	M	34	Scalp	5	10	Excision	No recurrence	12
		M	72	Scalp	5	2	Excision	No recurrence	8
		F	73	Scalp	2	6	Excision	No recurrence	6
Mori et al (18)	1	F	58	Scalp	5	10	Excision	Distant metastases; DOD	5
Park et al (19)	1	M	32	Scalp	2	10	Excision + RT	Distant metastases Lost to follow-up	63
Sau et al (20)	10	NS	NS	NS	NS	NS	Excision	Lymph node metastasis in one patient	NS
Seema et al (21)	1	F	60	Scalp	5	4	Excision	Lost to follow up	NA
Saida et al (22)	1	M	47	Scalp	8	10	Excision + RT	Lymph node metastasis	32
Takenaka et al (8)	1	F	59	Scalp	12	15	Surgery + RT + CT Ethanol injection	DOD (brain invasion), No metastases	18
Takata et al (23)	1	F	77	Scalp	4.5	15	Excision	No recurrence	84
Vandeweyer et al (24)	1	M	79	Ear	1	2	Excision	No Recurrence	18
Weiss et al (25)	1	M	78	Scalp	10	20	CT+ Excision	Lymph node metastasis	5
Waligora et al (26)	1	M	76	Scalp	11	15	Excision + RT	Lymph node metastasis	7, 5
Warner (27)	1	F	70	Scalp	8	2	Excision	No recurrence	15
Yoleri et al (28)	1	M	64	Scalp	7	30	Excision + RT	Soft tissue metastases;	8
Fillipou et al (29)	1	F	54	Scalp	NS	NS	Excision	No recurrence	24
Methis et al (30)	1	F	51	scalp	1	6	Excision	No recurrence	NS

CT: Chemotherapy, DOD: Died of disease, NA: Not available, NS: Not stated, RT: Radiotherapy

study the metastatic characteristics of this rare tumour.

The therapeutic approach in malignant proliferating trichilemmal cyst is the same as other malignant skin lesions. Adequate surgical excision remains the mainstay of treatment. The patient should be followed closely after surgery. The efficacy of alternative treatments for malignant cases cannot be safely evaluated in view of the very small number of published cases.

Only 39 well-documented cases of malignant proliferating trichilemmal cyst have been published to date in the English language literature. Table 1 summarizes their clinico-pathological features. There were 27 female and 11 male patients, ranging in age from 32 to 97 years. The most common site involved was the scalp. Two patients showed groin involvement and in another three patients, the ear, neck and lip was involved. Site was not specified in 10 patients. Surgical excision was done in all the patients. Combined radiotherapy and/or chemotherapy were also given in ten patients. Ten tumours showed metastasis: five to regional lymph nodes, three to distant sites and two to adjacent soft tissues. Takenaka *et al*[8] also demonstrated intra-tumoral ethanol injection as an alternative means of reducing tumour mass. On further analysis of data available, no conclusion can be derived regarding the optimal treatment of malignant proliferating trichilemmal cyst.

In summary, malignant proliferating trichilemmal cyst poses a diagnostic dilemma for the surgeon as well to the pathologist. Wide surgical excision should be considered as the primary modality of treatment while alternative therapies require further evaluation.

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