Case Report

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Surgery Section

Malignant Proliferating Trichilemmal Tumour of the Deltoid Region in an Adolescent: A Rare Presentation at an Unusual Site and Young Age

ADITYA CHADALAWADA[1], A NAVEEN KUMAR[2]

ABSTRACT

Proliferating Trichilemmal Tumour (PTT) is a rare benign adnexal neoplasm arising from the outer root sheath of the hair follicle. Malignant transformation to Malignant PTT (MPTT) is exceptionally uncommon and is frequently mistaken for squamous cell carcinoma, as the two share overlapping clinical and microscopic features. MPTT classically affects the scalp of women in the sixth-to-seventh decade of life; presentation in adolescence and at an extra-scalp site is rare. The authors report a 16-year-old girl with a painless swelling over the left deltoid region present for one year, with rapid enlargement over the preceding five months. There was no history of trauma or intramuscular injection. An outside incisional biopsy reported MPTT and she was referred for definitive management. Magnetic Resonance Imaging (MRI) showed a 3.1×2.5 cm subcutaneous mass superficial to the deltoid without muscular invasion and a few small left-axillary nodes. After tumour board discussion she underwent wide local excision with 1 cm margins and sentinel lymph node biopsy localised with methylene blue dye; the sentinel node was negative on frozen section and definitive histopathology and all resected margins were free of tumour. Histopathology confirmed MPTT with abrupt keratinisation, nuclear atypia, infiltrative margins and eight mitoses per 10 high-power fields, without lymphovascular invasion. She recovered well, did not require adjuvant radiotherapy and showed no recurrence at follow-up. The present case highlights that MPTT can occur in adolescents and at unusual sites. It must be distinguished from squamous cell carcinoma and complete surgical excision with margin assessment is the cornerstone of management.

Keywords: Adnexal and skin appendage neoplasms, Hair follicle, Outer root sheath, Surgical margins of excision

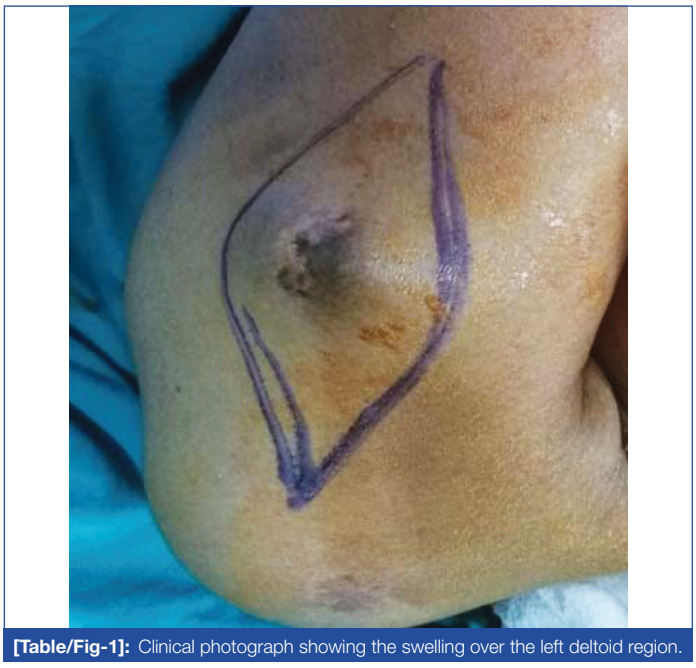
CASE REPORT

A 16-year-old girl presented with a painless swelling over the left deltoid region that had been present for one year and had gradually increased in size over the preceding five months. She was otherwise healthy with no significant past medical history. There was no history of trauma, intramuscular injection, or any local instrumentation at the site.

An incisional biopsy had been performed at an outside hospital on 14 March 2017 (reported 16 March 2017), which revealed a MPTT; definitive treatment was not undertaken there and she was referred to this tertiary surgical-oncology unit for staging and definitive management.

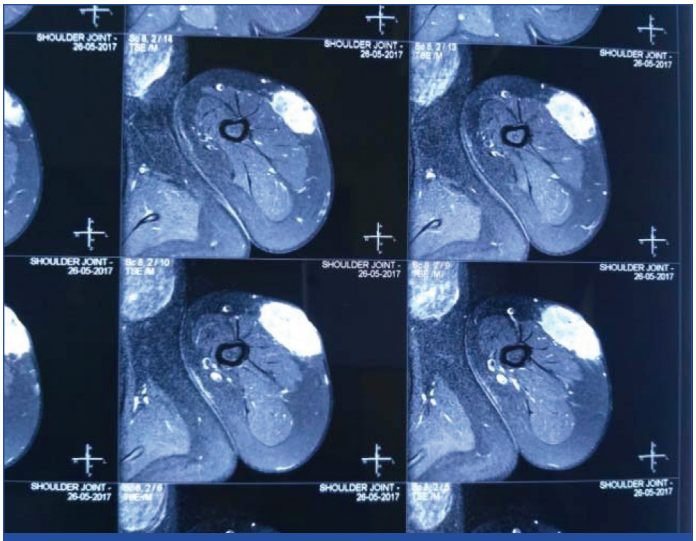
On arrival, the patient was haemodynamically stable. On examination there was a non tender, firm, nodular swelling measuring 3×3 cm over the left deltoid region [Table/Fig-1]. No axillary lymph nodes were palpable. Magnetic resonance imaging of the left shoulder [Table/Fig-2] showed a well-defined soft-tissue mass measuring 3.1×2.5 cm in the left arm, superficial to the deltoid and confined to the subcutaneous plane without muscular invasion. The lesion showed intense contrast enhancement with cystic areas and a few enlarged left-axillary lymph nodes were noted, the largest measuring 1.3×1.1 cm. There was no demonstrable pathology in the left humerus or brachial vessels and the visualised lung fields were clear.

The case was discussed at the tumour board and surgery was planned. She underwent wide local excision of the tumour with 1 cm margins all around and sentinel lymph node biopsy. The sentinel node was localised intraoperatively using methylene blue dye injected over the lesion; through a separate axillary incision the blue-stained sentinel node was identified and dissected. Intraoperatively the specimen margins were sent for frozen-section analysis and confirmed to be adequate. Frozen-section analysis of the sentinel lymph node was negative for malignancy, so axillary lymph node dissection was not performed. Operative findings confirmed no intramuscular extension.

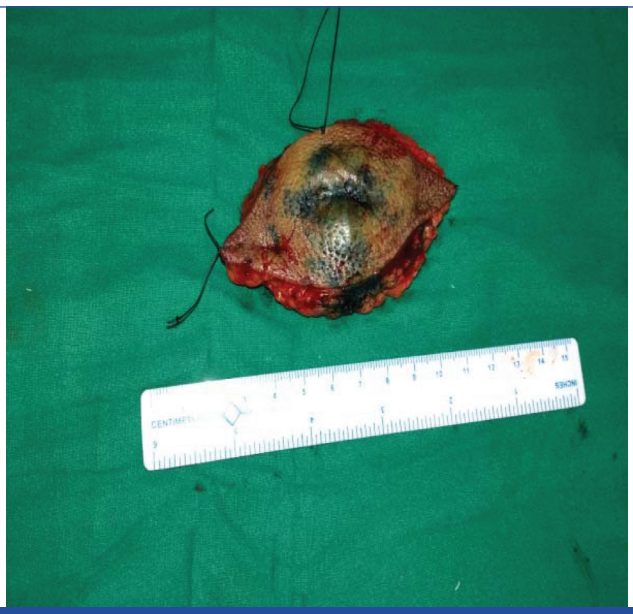


[Table/Fig-1]: Clinical photograph showing the swelling over the left deltoid region.

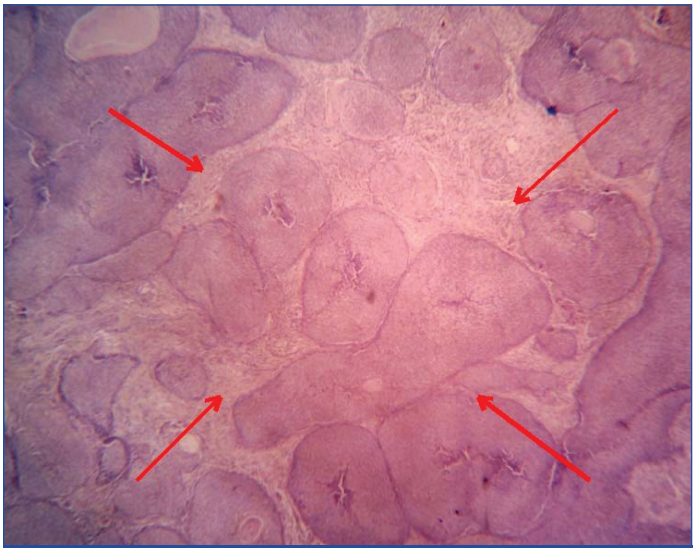
On gross examination the specimen was a lobulated subcutaneous mass measuring 3.5×3×2 cm [Table/Fig-3]. On histopathological examination, stratified squamous epithelium overlay a malignant neoplasm composed of nests and strands of atypical squamoid cells showing mild-to-moderate atypia with pleomorphism and hyperchromasia [Table/Fig-4]. Central keratin material with abrupt



[Table/Fig-2]: MRI of the left shoulder joint demonstrating a well-defined soft-tissue mass superficial to the deltoid muscle, confined to the subcutaneous plane.



[Table/Fig-3]: Gross specimen of the wide local excision with a margin greater than 1 cm all around (ruler for scale).



[Table/Fig-4]: Photomicrograph showing a neoplasm with nests of squamoid cells, central keratin material and abrupt keratinisation typical of MPTT. (Arrows indicate the abrupt keratinisation and infiltrating squamoid nests) (H&E, x40).

keratinisation was present and tumour cells were seen invading the surrounding stroma. Mitotic figures numbered eight per 10 high-power fields, no lymphovascular emboli were identified and all resected margins were free of tumour invasion (closest margins: superior 1.0 cm, inferior 1.5 cm, posterior 1.0 cm, anterior 0.5 cm, deep 0.8 cm and 0.2 cm from skin). These features were diagnostic of a MPTT.

As MPTT lacks a dedicated Tumour, Node, Metastasis (TNM) staging system, the lesion was graded histologically using the Ye J et al., classification as Group-3 (frankly malignant); clinically it represented a localised cutaneous adnexal malignancy that was completely excised with clear margins, sentinel-node negative (pN0), with no distant metastasis (M0) [1].

She was discharged on postoperative day 3. After consultation with a radiation oncologist, postoperative radiotherapy was not considered necessary. This decision followed multidisciplinary discussion and reflected the complete excision with histologically clear margins, a tumour-negative sentinel lymph node and the absence of regional or distant metastasis. She was reviewed in the outpatient clinic at 15 days for postoperative wound assessment and again at three months and six months. At each visit there was no clinical or radiological evidence of local recurrence, regional lymphadenopathy, or distant metastasis.

DISCUSSION

Skin tumours arising from the outer root sheath of hair follicles with trichilemmal-type keratinisation include the trichilemmal cyst, the PTT and its MPTT. PTT is an uncommon, usually benign, well-circumscribed subcutaneous lesion that can closely mimic squamous cell carcinoma both clinically and microscopically; it is also known as proliferating trichilemmal cyst or pilar tumour of the scalp [2]. It was first described by Wilson-Jones in 1966 as a proliferating epidermoid cyst and in 1995 it was recognised as an entity distinct from the proliferating epidermoid cyst [1]. Around 90% of PTTs arise on the scalp, with rarer reports on the forehead, nose, back, chest, abdomen, buttocks, elbow, wrist and vulva [3,4]. Most patients are women, typically in the sixth-to-seventh decade [5].

Malignant transformation is rare and is often heralded by sudden rapid growth. Histologically, MPTT shows marked nuclear atypia, cellular pleomorphism, atypical mitoses, dyskeratotic cells and infiltrative margins [6]. In the present patient, a year-long swelling that enlarged rapidly over five months raised suspicion of malignancy and microscopy confirmed it through infiltrative borders, nuclear atypia and increased mitotic activity (8 per 10 HPF). Ye J et al., proposed a useful histological grouping (benign, low-grade and frankly malignant) that helps distinguish MPTT from squamous cell carcinoma; the present case corresponds to the frankly malignant group [1,7]. Headington introduced the term “malignant proliferating trichilemmal cyst” for proliferating trichilemmal cysts undergoing malignant change; its true incidence is unknown owing to rarity and frequent misclassification as squamous cell carcinoma [8]. Histological features that favour MPTT over conventional squamous cell carcinoma include lobulated masses arising from the outer root sheath, abrupt trichilemmal-type keratinisation without an intervening granular layer and a continuous spectrum from benign to frankly malignant change; careful assessment of resection margins, mitotic rate and infiltrative growth helps to confirm frank malignancy and to guide management.

Two features make the present case noteworthy. First, the young age: MPTT in adolescents is exceptional, with most series reporting a mean age in the seventh decade; one of the youngest well-documented cases was a 26-year-old woman [9]. Second, the deltoid (extra-scalp) location, since the great majority of cases is scalp lesions. Recent reports have documented MPTT at other unusual sites - the clavicular region [10], the auricle [11], the thumb [12] and the neck with nodal metastasis [13] - and have emphasised aggressive or recurrent behaviour requiring multimodality treatment [14]. The present case adds an adolescent deltoid presentation that was localised, node-negative and completely resected.

The management of MPTT mirrors that of other malignant skin tumours: wide local excision with clear margins is the treatment of choice, with sentinel lymph node assessment helpful for nodal staging in suitable cases. Radiotherapy has been reported as an alternative for unresectable or refractory tumours [14]. Long-term follow-up is essential because the rarity of the disease precludes firm conclusions about adjuvant therapy. Adjuvant radiotherapy is not standardised and has generally been reserved for unresectable, recurrent, or incompletely excised tumours and for cases with other high-risk features, with the available evidence limited to case reports and small series; it was therefore not used in the present patient, who had a completely excised, node-negative lesion. Scheduled reviews at 15 days, three months and six months showed no recurrence, a pattern consistent with the close early surveillance advised after complete excision of MPTT; longer-term follow-up is being continued because late recurrence and metastasis are recognised.

CONCLUSION(S)

The MPTTs are rare and diagnostically challenging because they closely resemble squamous cell carcinoma and may therefore be under-reported. The present case underscores that MPTT can occur in adolescents and at unusual extra-scalp sites such as the deltoid region. Wide surgical excision with histologically clear margins remains the primary treatment, while the role of adjuvant therapy requires further study.

Author Contributions: Both authors managed this patient as resident surgeons in the same surgical unit; the affiliations shown reflect their current institutional positions. AC performed the clinical evaluation and perioperative management, collected the data, reviewed the literature and drafted and critically revised the manuscript. ANK contributed to the surgical planning and operative management, interpreted the imaging and histopathology findings and critically reviewed and edited the manuscript. Both authors approved the final version and agree to be accountable for all aspects of the work.

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PARTICULARS OF CONTRIBUTORS:
1. Senior Resident, Department of Gastrosurgery, Nizam Institute of Medical Sciences, Hyderabad, Telangana, India.
2. Consultant, Department of General Surgery, Srikara Hospitals, Peerzadiguda, Hyderabad, Telangana, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Aditya Chadalawada,
Bhel MIG 855, RC Puram, Hyderabad-502032, Telangana, India.
E-mail: adi9025338@gmail.com

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