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Navigating diagnostic complexity: A case of giant cell tumour of tendon sheath of thumb in a patient with prior hypopharyngeal Carcinoma

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Abstract
Giant Cell Tumour (GCT) is a benign neoplasm often found in the tendon sheaths or joints of the hands and feet. Its diagnosis can be challenging due to potential confusion with other benign soft tissue masses or even rare cases of metastasis. In this report, we present a case of a 65-year-old male with a history of post-chemo-radiotherapy-treated carcinoma (CA) of the hypopharynx, who was diagnosed with a Giant Cell Tumour (GCT) of the flexor tendon sheath of the right thumb. Diagnostic confusion initially arose between GCT and fibroma, epidermoid cyst, or acral metastasis due to the multinodular clinical appearance and the patient's history of malignancy. Following comprehensive radiological investigations, the patient underwent excision biopsy. The postoperative period was uneventful, leading to a stable discharge. This report discusses the clinical presentation, diagnostic challenges, course of investigation, diagnosis, and management of the case.

Keywords: Tenosynovial giant cell tumour, carcinoma hypopharynx, excision biopsy

Introduction
Tenosynovial Giant Cell Tumour (TGCTs) ranks as the second most common tumour affecting the hand after ganglion cysts [1]. It predominantly arises from the flexor tendons but can also occur on the extensor aspect of fingers, originating from the synovium of interphalangeal joints. Although less common, it may also appear in the ankle and toes. The condition primarily affects women in their third and fourth decades of life. Etiologically, TGCTs is associated with various factors including trauma, chronic inflammation, metabolic diseases, and potentially neoplastic processes. Clinically, it manifests as painless multinodular swellings typically located over the synovial lining of tendon sheaths. Bilateral involvement is frequently observed.
Despite being benign, TGCTs has a notable propensity for local recurrence following surgical excision, reported in up to 45% of cases [11]. The prevailing hypothesis regarding its pathogenesis suggests reactive or regenerative hyperplasia triggered by underlying inflammatory stimuli.
Treatment typically involves surgical excision aimed at complete removal of the tumour, often supplemented by radiotherapy in cases where there is concern for recurrence or incomplete resection. This combined approach has generally shown good outcomes in managing TGCTs, ensuring minimal impact on hand function and reducing the risk of recurrence.

Case presentation
The case involves a 65-year-old male patient with a known history of carcinoma of the hypopharynx, diagnosed two years ago. He underwent chemotherapy and radiotherapy for the condition. The patient presented to the outpatient department with a painless swelling on the anterior aspect of the pulp of the right thumb, which had been gradually increasing in size over the past eight months. There was no history of trauma, similar swellings in the past, positive family history, recent weight loss, or loss of appetite.
Physical examination revealed a 1 × 1 cm firm, nodule over the palmar aspect of the distal phalanx of the right thumb. The patient's finger mobility was preserved, and there were no sensory or motor deficits in the hand. Additionally, there was no muscle wasting and no similar swellings were noted in other areas.

Blood investigations, including white cell count and inflammatory markers such as erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP), were within normal limits.

An X-ray of the hand showed a soft tissue swelling with minimal erosion of the distal phalanx of the thumb (Fig. 1). Ultrasound of the right thumb demonstrated a small, lobulated, solid-appearing lesion along the volar aspect at the level of the distal phalanx. MRI revealed a cystic lesion measuring 10 × 0.5 × 10 mm on the flexor aspect of the distal phalanx of the thumb, which was iso-intense on T1-weighted images and hyper-intense on T2-weighted and STIR images, suggesting a benign lesion (Fig. 2).

Based on these findings, we considered a differential diagnosis of benign lesions, including an epidermoid cyst, fibroma, giant cell tumour of the tendon sheath (GCTTS), soft tissue neoplasm, or, rarely, acral metastasis.

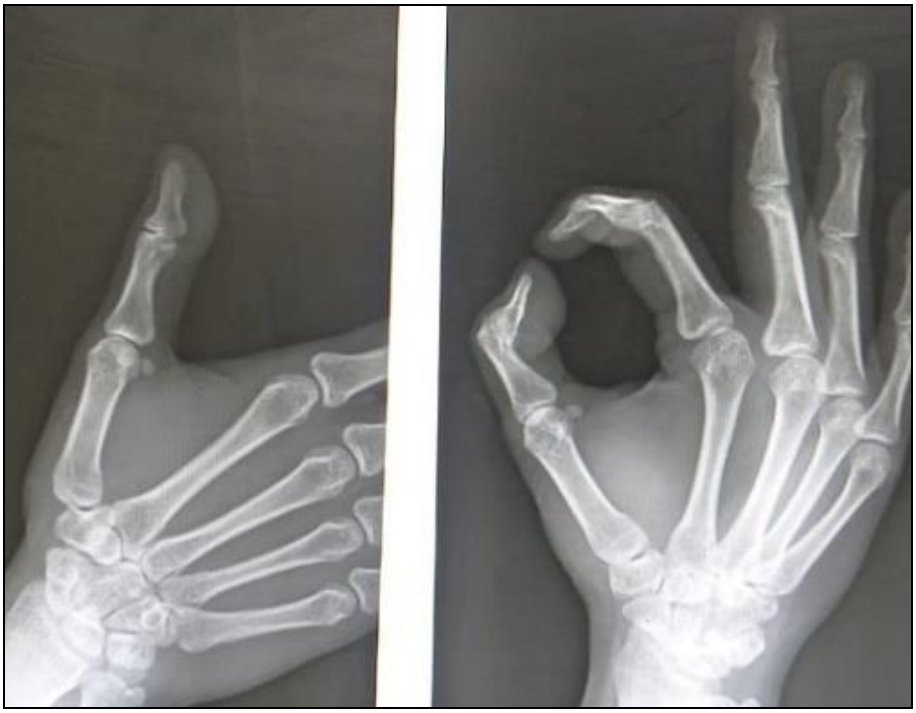

Fig 1: Radiographs of right hand

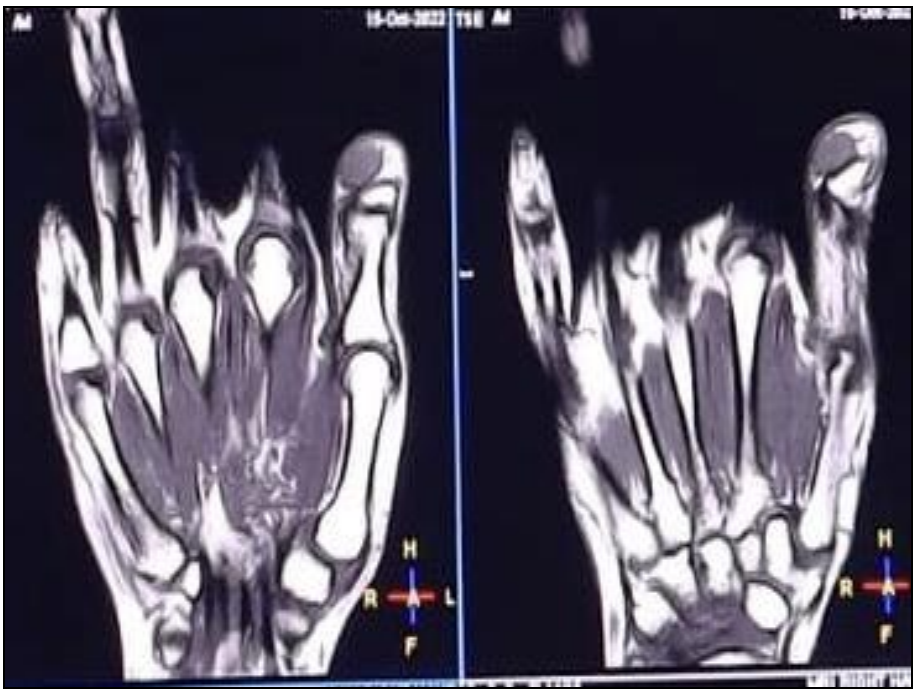

Fig 2: MRI showing cystic lesion measuring 1 on the flexor aspect of the distal phalanx of the thumb

An excisional biopsy of the swelling was performed (Fig. 3). The mass was found to be a yellowish, firm, nodular growth arising from the flexor pollicis longus tendon, with extension to the ulnar and dorsal aspects of the thumb.

Histopathological examination revealed tissues consisting of closely packed polyhedral cells with abundant hemosiderin-laden macrophages and foamy histiocytes, admixed with giant cells. There were also areas of hyalinization and

fibroblastic proliferation, suggestive of a Giant Cell Tumour (GCT) (Fig 4).

Following the excision biopsy, the postoperative period was uneventful, and the patient experienced a good recovery, regaining full movement of the thumb. There were no signs of recurrence observed after 6 months of follow-up.

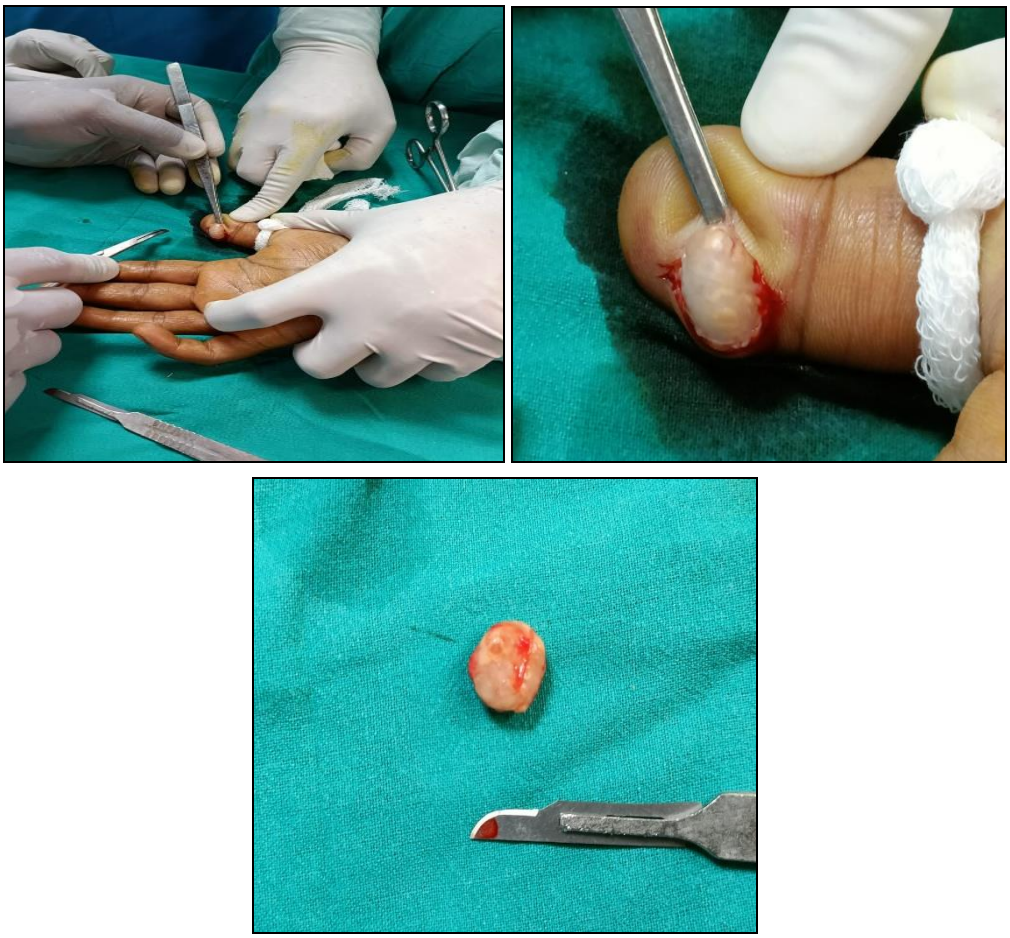

Fig 3: Intraoperative pictures

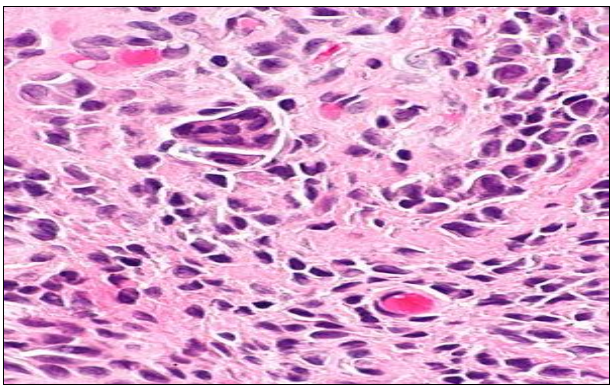

Fig 4-H and E: Staining showing tissues consisting of closely packed polyhedral cells with abundant hemosiderin-laden macrophages and foamy histiocytes, admixed with giant cells.

Discussion

We diagnosed our case as Tenosynovial Giant Cell Tumour (TGCT) of the right thumb. Upon presentation, following a detailed history and clinical examination, several differential diagnoses were considered. These included epidermoid cyst, fibroma, GCT, and acral metastasis from a primary malignancy, after ruling out other possibilities. Given the patient's history of Carcinoma of the hypopharynx diagnosed two years ago, and subsequent chemotherapy and radiotherapy, the possibility of acral metastasis or a malignant form of Tenosynovial GCT was also included in the differentials. Suzuki *et al*. [4] reported an 18% incidence of distant metastasis to bony extremities in patients with head-and-neck malignancies. Therefore, the potential for acral malignancy should be carefully considered in this patient with hypopharyngeal carcinoma.

Tenosynovial Giant Cell Tumours (GCT), of unknown etiology excluding ganglia, constitute nearly half of all soft tissue tumours in the hand. These tumours typically present as nodular forms and are commonly observed in patients aged 30 to 50 years, although they can occur across a wide age spectrum, predominantly affecting females (55% to 70% of cases). They are frequently located on the volar or latero-volar aspects of the fingers, particularly near the distal interphalangeal joint, without specific preference for any particular finger [1-3]. In terms of laterality, these lesions are more frequently found on the right hand and often affect the dominant hand [5, 7].

Recent studies have shown that cytogenetic abnormalities, such as trisomy 7 and autonomous growth, along with clinical features like local recurrence and case reports of metastatic GCTTS, suggest that it may be cancerous. Common genetic alterations in TGCTs include NOTCH1 mutations, NOTCH1 missense mutations, TP53 c.217-c.1178 missense mutations, TP53 R175H mutations, CSF-1 translocations, and loss of the CDKN2A/B gene.

Additionally, the patient had undergone chemo radiation for the management of hypopharyngeal cancer, which increased his risk for genetic mutations.

Though the suspected diagnosis of Tenosynovial Giant Cell Tumour (TGCT) of the hand is usually made clinically and through complementary imaging tests, including simple radiographs and ultrasound/MRI, which are typically sufficient, our case required confirmation through excisional biopsy. Clinically, TGCT is suspected when there is a solitary, soft tissue nodule that respects the skin, is often eccentric, painless, and slow-growing. Between 1% and 24% of patients report some pain, usually in conjunction with increased volume and sometimes neurovascular compromise [1, 5, 7].

The clinical differential diagnosis for TGCT is extensive. Ultrasound typically reveals a generally homogeneous, hypo- or hyperechoic solid lesion or, rarely, a heterogeneous one, with increased vascularity on Doppler study [7]. The lesion is associated with the tendon sheath, with which it does not move. It is also common to find satellite lesions. Although this lesion could be mistaken for ruptured synovial cysts, such confusion has no therapeutic significance if intervened upon.

Among complementary imaging techniques, MRI is considered by some to be the most accurate test for diagnosis. MRI can provide detailed images that help differentiate TGCT from other soft tissue masses, making it a valuable tool in the diagnostic process. By highlighting the tumor's relationship with adjacent structures, MRI aids in planning surgical excision if necessary.

The treatment of Tenosynovial Giant Cell Tumour (TGCT) is primarily based on the complete surgical removal of the lesion. This approach aims to prevent recurrence and is deemed more reliable when performed with a large exposition and the use of magnifying glasses or microsurgical tools, which allow for precise excision of the tumour. In our case the swelling was excised completely and there was no features of recurrence after 6 months follow up.

Radiation therapy, although controversial, can be considered for cases with a higher risk of recurrence. This includes lesions that exhibit significant mitotic activity, have bone involvement, or were not completely resected. In such cases, radiation therapy may help reduce the likelihood of the tumour returning. Recurrence can occur in up to 45% of treated cases, highlighting the importance of thorough initial treatment and consideration of adjuvant therapies.

Research by Wright has indicated that recurrences are more likely in highly cellular lesions with an increased number of mitoses [8]. Similarly, Rao and Vigorita observed that finding three or more mitotic figures in each high-power field could signify an actively growing lesion that is prone to recurrence [9]. These findings underscore the need for careful pathological examination and consideration of the tumour's cellular characteristics when planning treatment and follow-up care.

Recent studies have also suggested the potential role of the Nm23 gene as a prognostic marker for recurrence risk. Nm23 is a gene present in normal cells that is responsible for regulating cell infiltration and metastasis. Elevated levels of Nm23 in TGCTs may indicate a higher risk of the tumour recurring after treatment. This genetic marker could be used to identify patients who might benefit from closer monitoring and additional therapeutic interventions [10].

Conclusion

Tenosynovial Giant Cell Tumours (TGCTs) present predominantly as benign lesions, yet their diagnosis and management constitute a collaborative effort between surgeons and pathologists. Timely intervention is crucial, as any delay or misdiagnosis can evoke understandable anxiety in patients. Preoperative counselling regarding the potential for high recurrence rates is imperative for informed decision-making.

While malignant forms of TGCTs are rare, the management of these tumours necessitates a comprehensive approach integrating surgical precision, meticulous pathological evaluation, and consideration of adjunctive therapies such as radiation. Ongoing advancements in our understanding of the genetic and cellular underpinnings of TGCTs continually refine treatment strategies, promising enhanced outcomes and quality of life for patients.

In summary, the multidisciplinary management of TGCTs underscores the importance of proactive diagnosis, patient-centered care, and ongoing research to optimize therapeutic efficacy and mitigate recurrence risks in this complex spectrum of conditions.

Abbreviations

TGCT: Tenosynovial Giant Cell Tumour
GCT: Giant cell tumour
MRI: Magnetic resonance imaging

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Conflict of interest

No conflict of interest.

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Informed consent

Written informed consent obtained for publication of data, images and treatment related documents.

Author's contributions

All Authors contributed equally to conceptualization, validation, visualization, writing- review and editing.

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