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TITLE: AN UNUSUAL CASE OF LEFT LIMB HYPERTROPHY: A CLINICAL EXPLORATION

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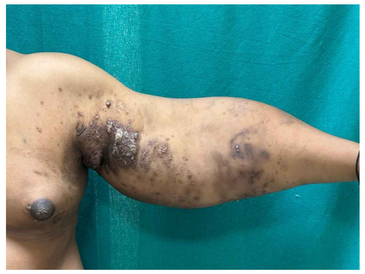
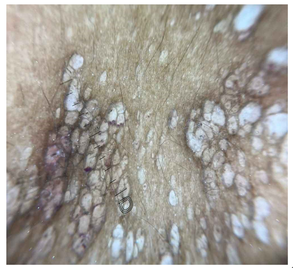
Introduction

Klippel–Trenaunay syndrome (KTS) is a rare vascular anomaly within the PIK3CA-related overgrowth spectrum (PROS), classically presenting with the triad of capillary malformations, venous abnormalities, and soft tissue or bony hypertrophy. Although lower limb involvement is most common, upper limb presentation is uncommon and can pose diagnostic and therapeutic challenges.

Case presentation

A 15-year-old female presented with progressive hypertrophy of the left upper limb since birth, associated with a port-wine stain extending to the axilla. Examination revealed limb enlargement in both length and girth, prominent superficial veins and varicosities, and multiple papular lesions with intermittent serous discharge in the axilla. The skin was thickened, warm, and showed hyperpigmentation and crusting, though no bruit, pulsation, ulceration, or active infection was detected. Based on the coexistence of capillary malformation, venous anomalies, and soft tissue hypertrophy, a clinical diagnosis of KTS was established.



Examination findings

- Left upper arm is 80cms and right upper arm is 21cms
- On palpation, no pulsation noted
- On auscultation, no bruit noted
- Dermoscopy reveals numerous white-to-yellowish lacunae, consistent with dilated lymphatic spaces. Areas of background erythema and subtle telangiectasias are observed.

Investigations

- Routine laboratory investigations were within normal limits
- Color Doppler of the left upper extremity for venous blood flow reveals features of AVM in the subclavian & axillary veins
- MRI shows slow-flow AVM in the left arm with extension into the left lateral chest wall
- Biopsy shows squamous epithelium and a few dilated lymphatics beneath it, suggestive of lymphangiectasia
- PIK3CA gene mutation: Reports awaited

Management

Management is multidisciplinary, involving dermatology, interventional radiology, and vascular surgery. Symptomatic strategies include compression therapy, sclerotherapy, and laser ablation. Sirolimus has recently shown promise, particularly in cases with lymphatic involvement.

Conclusion

This case highlights an unusual presentation of KTS in the upper limb, emphasizing the importance of recognizing atypical disease patterns. Individualized, multidisciplinary management is essential for optimal outcomes in patients with PROS disorders.

References

1. John PR. Klippel-Trenaunay Syndrome. Tech Vasc Interv Radiol. 2019;22(4):100634. doi:10.1016/j.tvir.2019.100634.
2. Lamar LM, Farber GA, O'Quinn SE. Klippel-Trenaunay-Weber syndrome. Arch Dermatol. 1965;91:58-9. doi:10.1001/archderm.1965.01600070064009.
3. Maharjan B, Liu L, Liu X, Liu Q, Jiang X. Klippel-Trenaunay syndrome in the distal part of the unilateral upper limb and venous deficiency: a case report. JNMA J Nepal Med Assoc. 2025;63(282):123-6. doi:10.31729/jnma.8892.
4. Asghar F, Aqeel R, Farooque U, Haq A, Taimur M. Presentation and management of Klippel-Trenaunay syndrome: a review of available data. Cureus. 2020;12(5):e8023. doi:10.7759/cureus.8023.