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**Case Report**

# Klippel-Trenaunay syndrome: As a hyperpigmented plaque in a Pakistani child

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**Abstract** Hailey-Hailey disease is an autosomal dominant acantholytic disorder relatively uncommon in India. It is characterized by painful, pruritic, foul smelling vesicles and bullous lesions with erosions in Klippel-Trenaunay Syndrome (KTS) is an extremely rare genetic condition marked by the presence of capillary malformations, abnormal overgrowth of soft tissues, along with venous malformations often involving one limb. We report a case report of a 14-years-old boy who had a hyperpigmented plaque on left leg, ultimately diagnosed with KTS. Despite the rarity of this syndrome, recent case reports from Pakistan highlight the challenges in diagnosis and management within the local healthcare context. Our case underscores the importance of awareness among healthcare providers regarding the clinical manifestations of KTS and the need for advanced imaging techniques for accurate evaluation. By sharing our experience, we aim to contribute to the understanding of KTS and improve patient care in Pakistan and similar resource-limited settings.

***Key words***
Klippel-Trenaunay syndrome; Vascular malformations; Hyperpigmented plaque.

## Introduction

Klippel-Trenaunay Syndrome (KTS) is an extremely uncommon genetic disorder marked by capillary malformations (like port-wine stains), overgrowth of soft tissue or bone, and venous malformations. This syndrome primarily affects one extremity, often the lower limb, and can lead to various complications, including vascular abnormalities and limb overgrowth.[1,2] While the exact prevalence of KTS remains uncertain, it is estimated to occur in approximately 1 in 100,000 live births.[3] Despite its rarity, KTS poses significant challenges in diagnosis and management, particularly in resource-limited settings such as Pakistan.

In Pakistan, limited literature exists regarding Klippel-Trenaunay Syndrome, with only a few reported cases documented in medical literature. However, recent advancements in diagnostic modalities and therapeutic approaches have facilitated the recognition and management of this complex condition.[4-8] Understanding the clinical presentation, diagnostic workup, and management strategies of KTS is crucial for healthcare providers in Pakistan to ensure timely intervention and improve patient outcomes. In this report, we present a case of Klippel-Trenaunay Syndrome in a 14-year-old boy from Pakistan, highlighting the clinical features, diagnostic approach, and management strategies employed in the local healthcare setting. By sharing our experience and insights, we aim to contribute to the growing body of knowledge on KTS and enhance the understanding of this rare syndrome among healthcare professionals in Pakistan and beyond.

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**Case report**

A 14-year-old male presented to the dermatology clinic with complaint of a hyperpigmented plaque on his left leg that had been present since birth, and had been progressively enlarging over the past few years. The lesion was asymptomatic and did not cause any pain or discomfort. At birth, the lesion was small and pink in colour on lower part of left shin, which gradually enlarged in size, and its colour changed to dark grey. The patient denied any history of trauma to the area. There was no associated bleeding, discharge, or pruritus. Over the years, prominent vessels started appearing on the leg around and under the lesion. There was a discrepancy in the girth of lower limbs, since the age of six months, due to which his gait was also unstable. He also expressed concern about the cosmetic appearance of the lesion. He was born to consanguineous parents and denied any family history of similar skin lesions.

Upon examination, a large hyperpigmented patch was noted on the lateral and posterior aspect of the patient's left lower limb, extending from the back of knee to the foot **(Figures 1, 2)**. The lesion had irregular borders and variegated pigmentation, ranging from light brown to dark brown. There were no signs of inflammation, ulceration, or induration. Palpation revealed a soft, non-tender, and non-pulsatile mass beneath the patch. Peripheral pulses were intact, and there were no signs of lymphadenopathy. Multiple varicosities were seen around the plaque. On further exposure, left leg was visibly larger than the right. There was a 5 cm difference in girth at the level of mid thigh, 2.2 cm at the level of mid-calf, while the difference was 3.8 cm at the level of ankle **(Figure 3)**.

Given the unusual appearance of the lesion and its characteristic distribution, further investigations were warranted. Plain radiograph of left leg showed soft tissue thickening extending from mid calf to ankle, having phleboliths at distal end near ankle joint. Underlying bones were normal. Doppler ultrasound of left leg was performed, revealing few varicosities along the course of short and great saphenous vein with a prominent perforator (diameter less than 3.5cm). Doppler ultrasound of right leg and abdomen were normal.

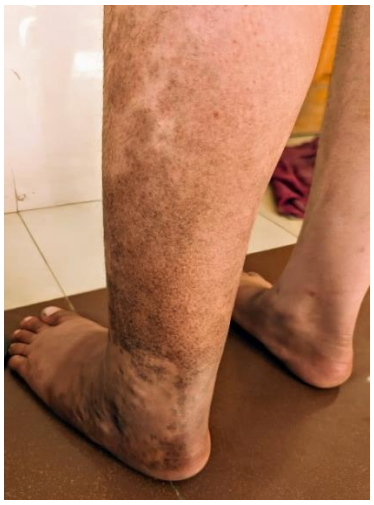

**Figure 1** Lateral aspect of left leg showing hyperpigmented patch along with varicosities on ankle and foot.

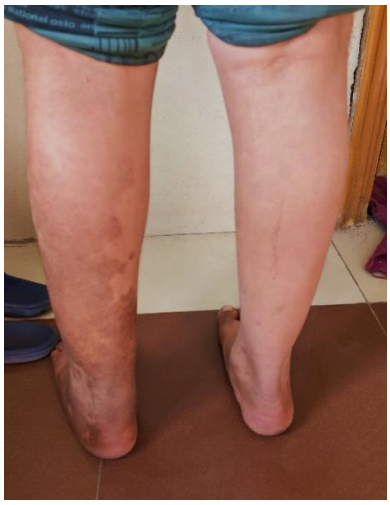

**Figure 2** Posterior aspect of left leg showing hyperpigmented plaque along with difference of girth between both legs.

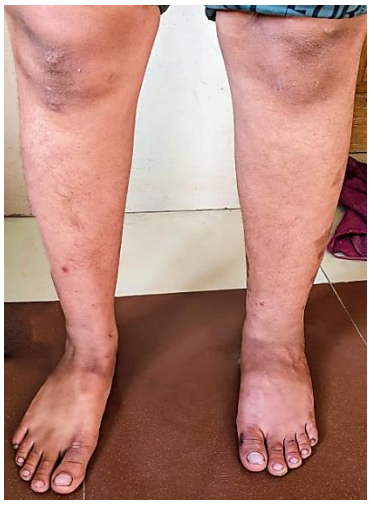

**Figure 3** Anterior aspect of both legs showing difference of girth.

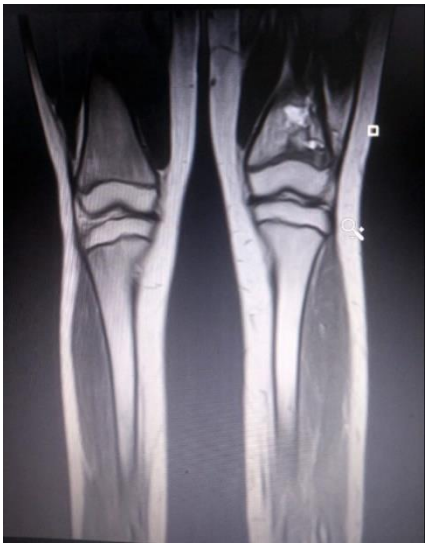

**Figure 4** Computed tomography angiogram of legs showing soft tissue thickening of left leg along with vascular malformation involving lower end of left femur.

Computed tomography (CT) angiogram was also conducted to assess the extent of the vascular abnormalities and rule out associated complications such as deep vein thrombosis. It showed soft tissue hypertrophy of left leg compared to right **(Figure 4)**. There was mild skin thickening with adjacent tiny vessels (capillaries malformation) on anterolateral aspect of left leg. Dilated tortuous venous channels were noted along the lateral aspect of left leg infiltrating deep compartment of leg. In arterial phase, multiple prominent venous channels were appreciated in soft tissue, suggesting arteriovenous malformation. However, no definite high flow arteriovenous shunting was appreciated. No evidence of deep vein thrombosis or lymphatic malformation was noted. Bone window images showed trabeculated heterogeneous lesion in left femur consistent with intraosseous involvement due to vascular malformation **(Figure 4)** which was confirmed on Magnetic resonance imaging (MRI) as well.

Based on the clinical presentation and imaging findings, a diagnosis of Klippel-Trenaunay Syndrome was established. The patient's hyperpigmented plaque was attributed to cutaneous vascular malformations, a common feature of KTS. The patient and his family were counseled regarding prognosis and therapeutic options and was referred to a vascular surgeon. However, they wanted to continue with conservative management.

## Discussion

Klippel-Trenaunay Syndrome typically affects one extremity, most commonly the lower limb, as observed in this case. The exact etiology of KTS remains unclear, but it is believed to result from somatic mutations in genes involved in vascular development.[1]

The clinical manifestations of KTS can vary widely, ranging from cosmetic concerns to severe complications such as thrombosis, bleeding, and limb overgrowth. Management strategies focus on addressing specific symptoms and may include conservative measures such as compression therapy, laser treatment for cutaneous lesions, and surgical intervention for vascular malformations or limb discrepancy.[3]

## Conclusion

This case highlights the importance of recognizing the cutaneous manifestations of Klippel-Trenaunay Syndrome, particularly in pediatric patients presenting with hyperpigmented plaques or vascular abnormalities. Early diagnosis and appropriate management are essential to mitigate potential complications and improve the patient's quality of life.

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***Declaration of patient consent*** Written informed consent was obtained from the parents of the patient for the publication of this case report and accompanying images..

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***Conflict of interest*** The authors declare no conflicts of interest related to this case report.

***Authors' contribution***

**HT,II,MZ,FA**: Diagnosis and management of the case, critical review of the manuscript, has given final approval of the version of the manuscript to be published.

**SA,SB**: Identification and management of the case, manuscript writing, has given final approval of the version of the manuscript to be published.

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