ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.85.90

Journal of Dermatological Case Reports

Unilateral Hypertrophy and Bilateral Vascular Malformations in an Adolescent: A Case of Klippel-Trenaunay Syndrome

Dr Japsimran Singh Nagpal[1], Dr Shilpa S Haibati (Pathrikar)[2]
[1]Junior Resident Department of Dermatology MGM Medical college & Hospital, Chhatrapati Sambhajinagar,MGM Institute of Health Sciences, Navi Mumbai. japsimrannapal96@gmail.com
[2]Associate Professor Department of Dermatology MGM Medical college & Hospital, Chhatrapati Sambhajinagar,MGM Institute of Health Sciences, Navi Mumbai drshilpahaibatti@gmail.com

Corresponding Author

Dr Japsimran Singh Nagpal
Junior Resident Department of Dermatology MGM Medical college & Hospital, Chhatrapati Sambhajinagar,MGM Institute of Health Sciences, Navi Mumbai
japsimrannapal96@gmail.com

Abstract:

Introduction:
Klippel-Trenaunay Syndrome (KTS) is a rare, complex congenital disorder characterized by a triad of capillary malformations (port-wine stains), venous varicosities, and limb hypertrophy. While typically unilateral, phenotypic expression varies significantly. We present a case of an adolescent male exhibiting extensive cutaneous involvement and mixed vascular anomalies.
Case Presentation:
A 16-year-old male presented for evaluation of congenital vascular abnormalities. Physical examination revealed an extensive capillary malformation (port-wine stain) involving the left chest and abdomen, extending to the bilateral lower extremities. Significant varicosities were noted in the bilateral lower limbs, indicative of chronic venous insufficiency. Additionally, a distinct haemangioma was identified on the left ankle. Assessment of limb proportions demonstrated localised soft tissue and bony hypertrophy of the right foot, confirming the clinical triad characteristic of KTS. Diagnosis was clinical, supported by the presence of the defining triad. Doppler ultrasound was utilised to assess deep venous patency and characterise the extent of the varicosities and the ankle haemangioma. The patient's presentation aligns with the somatic mosaicism often associated with PIK3CA mutations, which can lead to tissue overgrowth and vascular dysplasia.
Management:
Management for this patient focused on symptom alleviation and prevention of complications. Conservative measures, including graduated compression therapy, were initiated to manage venous hypertension and oedema in the lower limbs. The patient is under multidisciplinary surveillance involving dermatology, vascular surgery, and orthopaedics to monitor the right foot hypertrophy and assess the need for potential sclerotherapy or epiphysiodesis (growth modulation) should functional impairment arise.
Conclusion:
This case highlights the heterogeneity of Klippel-Trenaunay Syndrome, particularly the presence of bilateral cutaneous and venous symptoms alongside unilateral hypertrophy. It underscores the necessity for thorough clinical evaluation and individualized, multidisciplinary care to optimize quality of life in adolescent patients transitioning to adulthood.

Keywords:

Klippel–Trenaunay syndrome; Vascular malformations; Port-wine stain; Limb hypertrophy; Varicosities; Doppler ultrasonography; Congenital vascular disorder

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.85.90

Journal of Dermatological Case Reports

Received : 26-04-2026 Revised : 01-05-2026 Accepted: 10-05-2026 Published : 13-05-2026

Introduction

Klippel–Trenaunay syndrome (KTS) is a rare congenital disorder involving vascular malformations, characterized by a classical triad of capillary malformations (port-wine stains), venous abnormalities, and hypertrophy of soft tissue and/or bone [1]. It usually occurs sporadically, with an estimated incidence of about 1 in 20,000–40,000 live births [2]. The clinical presentation varies greatly, from mild skin involvement to extensive vascular and limb anomalies that may cause significant functional impairment and complications [3].

KTS is now classified within the PIK3CA-related overgrowth spectrum (PROS), resulting from somatic activating mutations in the PIK3CA gene that cause abnormal cell growth, tissue enlargement, and vascular malformations [4]. Recent developments in vascular anomaly classification, especially by the International Society for the Study of Vascular Anomalies (ISSVA), have clarified that KTS is a low-flow vascular malformation involving capillaries, veins, and lymphatic vessels [5].

Although KTS usually involves a single limb, atypical cases, such as bilateral vascular malformations or disproportionate limb hypertrophy, have been observed, complicating diagnosis and treatment [6]. Besides skin and musculoskeletal issues, patients can experience complications such as chronic venous insufficiency, thromboembolic events, and, less frequently, visceral vascular involvement [7].
Management of KTS is primarily conservative and requires a multidisciplinary approach focused on symptom control, complication prevention, and quality-of-life enhancement. Treatment options include compression therapy, sclerotherapy, and surgical interventions in selected cases, with emerging evidence supporting targeted therapies such as sirolimus in complex vascular malformations [2].

This case describes an adolescent male with KTS presenting with bilateral vascular involvement and unilateral limb hypertrophy, emphasising the clinical variability of the condition and the need for individualized evaluation and management.

Case Report

A 16-year-old male visited the dermatology outpatient department with complaints of gradually worsening dilated veins in both lower limbs and a reddish skin discolouration that has been present since birth. The skin lesion first appeared on the left chest and abdomen and gradually spread to both lower limbs. There is no history of pain, ulceration, bleeding, or similar issues in the family.
During the clinical exam, a large, well-defined, non-blanchable port-wine stain was observed over the left chest and abdomen, extending to both lower limbs. Multiple dilated, tortuous superficial veins appeared on both legs, indicating venous malformations. Additionally, a soft, compressible vascular lesion consistent with a haemangioma was observed over the left ankle.

Further examination revealed that the right foot was disproportionately enlarged due to soft-tissue and bone hypertrophy, resulting in asymmetry. No major systemic abnormalities were found during general or systemic examinations.

Colour Doppler ultrasonography of the lower limbs showed dilated superficial veins with venous reflux. The deep venous system was open and compressible. No arteriovenous shunting was observed, ruling out high-flow vascular malformations. The lesion on the left ankle appeared to be a low-flow vascular lesion.

Based on the presence of capillary malformation, venous abnormalities, and limb hypertrophy, a

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.85.90

Journal of Dermatological Case Reports

clinical diagnosis of KTS was made and supported by radiological findings.
The patient was informed about the chronic and progressive nature of the condition. Conservative treatment was started, including graduated compression therapy to decrease venous stasis and oedema, along with advice on proper skin care to prevent complications like dermatitis and ulcers. The patient was also encouraged to have regular follow-up visits and multidisciplinary evaluations with dermatology, vascular surgery, and orthopaedics to monitor the condition and determine if additional treatment is needed.

Radiological Findings And Diagnostic Considerations

Colour Doppler ultrasound of the lower limbs revealed several enlarged superficial veins with venous reflux. The deep veins were normally patent and compressible. No arteriovenous shunting was detected, excluding high-flow vascular malformations. The vascular abnormality on the left ankle showed features typical of a low-flow lesion, suggesting a venous malformation.

Considering the combined clinical and radiological findings, a provisional diagnosis of a congenital vascular malformation syndrome was established. The primary differential diagnoses included Parkes–Weber syndrome, typically associated with high-flow arteriovenous malformations, and other overgrowth syndromes such as CLOVES and Proteus syndrome, which may present with asymmetric limb overgrowth and vascular anomalies. However, the absence of arteriovenous shunting on Doppler imaging and the presence of low-flow vascular malformations strongly favoured a diagnosis of KTS.
A notable feature in this case is the presence of bilateral venous malformations alongside unilateral right foot hypertrophy, representing an atypical presentation. The coexistence of capillary malformation, venous abnormalities, and localized limb overgrowth meets the classical diagnostic criteria of KTS.

Overall, the clinical and radiological findings align with Klippel–Trenaunay syndrome (KTS).

Diagnosis

The diagnosis for this patient was mainly clinical, supported by radiological evidence. It was confirmed by the classic triad of KTS, including capillary malformation (port-wine stain), venous abnormalities such as bilateral varicosities, and limb hypertrophy involving the right foot.

Management

The patient was counselled on the chronic and progressive nature of the condition. Conservative management was initiated, including graduated compression therapy to reduce venous stasis and associated symptoms. Additionally, appropriate skin care advice was given to minimise the risk of complications such as dermatitis and ulceration.
The patient was scheduled for regular follow-up visits, during which Doppler ultrasonography was used for radiological surveillance to monitor venous issues and identify possible complications. A multidisciplinary team was advised for continuous assessment and treatment.

Discussion

KTS is a rare congenital vascular malformation characterized by the classical triad of capillary malformations, venous abnormalities, and soft tissue or bony hypertrophy [1,2]. The clinical spectrum is highly variable, ranging from mild cutaneous involvement to extensive vascular anomalies with functional impairment [3,6].
The present case is notable for the coexistence of **bilateral venous malformations with unilateral hypertrophy of the right foot**, representing an atypical presentation. Similar variability has been reported in the literature, with KTS not always conforming to the classical unilateral pattern described by **Karmacharya et al. (2022)** [8].

Radiological evaluation plays a crucial role in diagnosis. In this case, Doppler ultrasonography demonstrated dilated superficial venous channels with reflux and a normal deep venous system, without evidence of arteriovenous shunting. These findings are consistent with

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.85.90

Journal of Dermatological Case Reports

low-flow vascular malformations, as defined by ISSVA classification [5,9].

The differential diagnosis includes Parkes–Weber syndrome, which is characterized by **high-flow arteriovenous malformations**, in **contrast to the low-flow pattern observed in KTS** [10]. Further imaging-based evaluation supports the distinction between low-flow and high-flow vascular anomalies [11].

Recent advances have improved understanding of vascular malformations and their management, including the role of targeted therapies, such as sirolimus, in complex cases [12]. In addition, variations in clinical presentation, including atypical distribution and disease extent, have been documented in multiple studies [13].

Patients with KTS are at risk of complications such as chronic venous insufficiency, thrombosis, and, less commonly, visceral involvement [7,14]. As highlighted by Reis et al. (2018) [7], thromboembolic complications are an important clinical concern that requires regular monitoring.
Overall, this case reinforces the importance of correlating clinical findings with imaging to establish an accurate diagnosis of KTS, particularly in atypical presentations. Recognition of such variants is essential to avoid misdiagnosis and to guide appropriate evaluation and management strategies.

Conclusion

KTS is an uncommon congenital vascular disorder with a wide spectrum of clinical presentations. This case highlights an unusual variant, with bilateral venous malformations and hypertrophy of one limb, underscoring the disorder's variability. Accurate diagnosis requires thorough clinical evaluation and imaging to differentiate KTS from other vascular malformation syndromes.
A multidisciplinary approach combined with continuous follow-up is essential for managing the condition effectively and preventing complications. Early diagnosis and personalised treatment are crucial for enhancing functional outcomes and quality of life in patients.

References

1. Klippel M, Trenaunay P. Du naevus variqueux osteohypertrophique. Arch Gen Med. 1900;3:641–672.
2. 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:e8023.
3. Martinez-Lopez A, Salvador-Rodriguez L, Montero-Vilchez T, Molina-Leyva A, Tercedor-Sanchez J, Arias-Santiago S. Vascular malformation syndromes: an update. Curr Opin Pediatr. 2019;31:747–753.
4. Vahidnezhad H, Youssefian L, Uitto J. Klippel-Trenaunay syndrome belongs to the PIK3CA-related overgrowth spectrum (PROS). Exp Dermatol. 2016;25:17–19.
5. International Society for the Study of Vascular Anomalies (ISSVA). ISSVA classification of vascular anomalies [Internet]. Available from: https://www.issva.org/classification
6. Karmacharya RM, Vaidya S, Bhatt S, et al. Klippel-Trenaunay syndrome: case series from a university hospital. Ann Med Surg (Lond). 2022;78:103732.
7. Reis J, Alomari AI, Trenor CC, et al. Pulmonary thromboembolic events in patients with Klippel-Trenaunay syndrome. J Vasc Surg Venous Lymphat Disord. 2018;5:511–516.
8. Sung HM, Chung HY, Lee SJ, et al. Clinical experience of Klippel-Trenaunay syndrome. Arch Plast Surg. 2015;42:552–558.
9. Schoch JJ, Nguyen H, Schoch BS, Anderson KR, Stans AA, Driscoll DJ, et al. Orthopaedic diagnoses in patients with Klippel-Trenaunay syndrome. J Child Orthop. 2019;13:457–462.
10. Redondo P, Aguado L, Martínez-Cuesta A. Diagnosis and management of extensive vascular malformations of the lower limb: Part I. Clinical diagnosis. J Am Acad Dermatol. 2011;65:893–908.
11. John PR. Klippel-Trenaunay syndrome.Tech Vasc Interv Radiol. 2019;22:100634.

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.85.90

Journal of Dermatological Case Reports

12. Hammer J, Seront E, Duez S, Dupont S, Van Damme A, Schmitz S, et al. Sirolimus is efficacious in treatment for extensive and/or complex slow-flow vascular malformations: a monocentric prospective phase II study. Orphanet J Rare Dis. 2018;13:191.
13. Cha SH, Romeo MA, Neutze JA. Visceral manifestations of Klippel-Trenaunay syndrome. Radiographics. 2005;25:1694–1697.
14. Tyagi S, Harshavardhan JKG. Exploring the complexity of Klippel–Trenaunay syndrome: a case study and review. J Ortho p Case Rep. 2025;15(8):265–269.
15. Rook's Textbook of Dermatology. 9th ed. Wiley-Blackwell; 2016.
16. IADVL Textbook of Dermatology. 4th ed. Bhalani Publishing House; 2016

Image 1:Extensive port- wine stain over left chest and abdomen

Image 2:Bilateral lower- limb varicosities with marked hypertrophy of the right foot.

ISSN : (Online): 3008-038X
Abbreviation: J Derm Cse Rep
DOI: 10.61705/jdcr.19.2.2026.85.90

Journal of Dermatological Case Reports

Image 3:Right lower limb showing prominent varicose veins and disproportionate soft- tissue/bony enlargement of the foot.