VOLUME - 13, ISSUE - 11, NOVEMBER - 2024 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

General Surgery

VEIN AND VIGOR: EXPLORING THE RARE KLIPPEL TRENAUNAY SYNDROME

Dr. R. Niranjan Kumar* Assistant Professor, Institute of General Surgery, Madras Medical College, Chennai, Tamil Nadu, India. *Corresponding Author

Dr. Alice. P Postgraduate, Institute of General Surgery, Madras Medical College, Chennai, Tamil Nadu, India.

ABSTRACT Klippel Trenaunay syndrome is a rare genetic vascular disorder characterized by a triad of varicose veins, capillary malformation and limb hypertrophy with an incidence of 1 in 100000[1]. The case report highlights the clinical presentation, diagnosis and management of a 28-year-old female presenting with dilated tortuous Vein of Servelle and the characteristic port-wine stain. This review aims to synthesize the current knowledge about the etiology, clinical features, diagnosis and newer treatment options for better patient care.

KEYWORDS : Klippel Trenaunay syndrome; vein of Servelle, port wine stain, hemangioma

INTRODUCTION:

Klippel Trenaunay syndrome was first described by French physicians Maurice Klippel and Paul Trenaunay in 1900[1]. The exact pathophysiology remains unclear. However, this rare disorder is found to have been related to genetic mutations in the PIK3CA gene which is responsible for the growth of cells and development of tissues in the body. This leads to activation of protein kinase PI3K and cell overgrowth by dysregulation of the mTORC2 pathway. [4] [5]. Recently KTS is grouped under PROS (PIK3CA related overgrowth syndromes). Occasionally translocations of chromosomes 5-11 and 8-14 have been reported. [5]

CASE REPORT:

A 28-year-old female cashier came to the outpatient department with chief complaints of dilated tortuous veins on her left leg for 13 years that has recently increased in size over the past one month. The swelling had progressed from below upwards and was associated with dull, aching, intermittent pain that aggravated on exertion.

She gave no history of rest pain or similar complaints in the past. There was no significant personal or family history.

Examination revealed multiple dilated tortuous Veins on the posterolateral aspect of the left leg extending up to the posterior middle thigh and a single engorged dilated vein of diameter two centimeters extending from posterior aspect of leg to mid-thigh. The left leg was comparatively larger than the right.

Characteristic port wine stain and multiple hemangiomas were noted on the lateral aspect of the left thigh and leg. Hyper pigmentation and venous eczema were found over the Gaiters area.

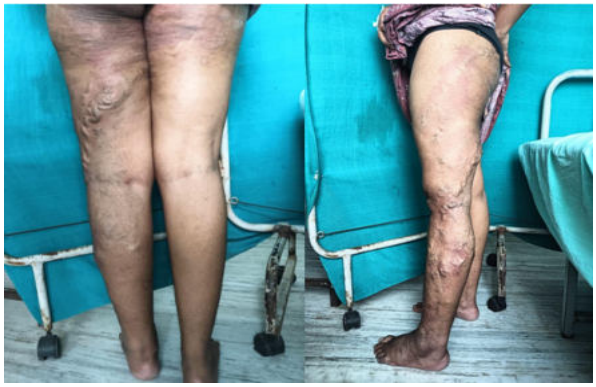

Fig 1: Clinical photograph of left lower limb Showing vein of Servelle and portwine stain

The following tests were done with inference as follows:
Trendelenburg I: Negative
Trendelenburg II: Positive
Morrissey's cough impulse test: Negative
Schwartz test: Positive over dorsovenous column
Multiple tourniquet test: Positive
Fegans palpation: Defects in deep fascia were palpable.

Bedside tests for DVT such as Modified Perthes test, Homans sign and Moses sign were negative.

Peripheral pulses of both upper and lower limbs were intact.

Radiograph of the limb showed soft tissue hypertrophy and vessel wall calcifications.

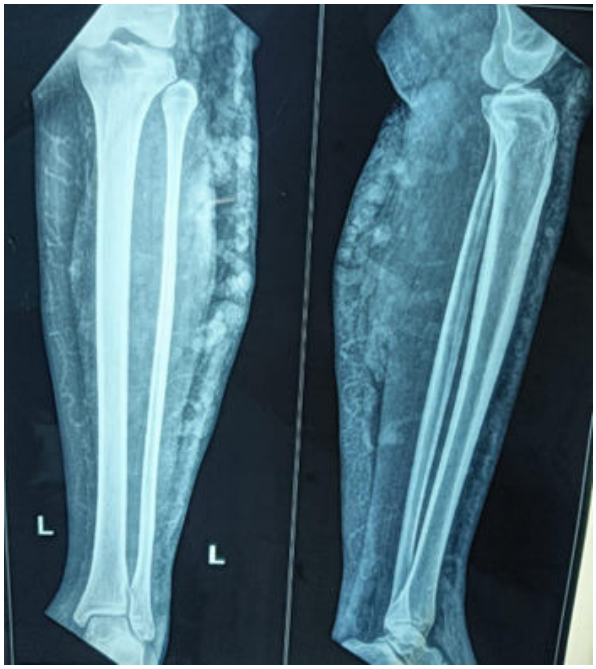

Fig 2: Radiograph of left leg showing soft tissue hypertrophy and vessel wall calcification.

DUPLEX scan confirmed perforator incompetence. Vein of Servelle or the lateral marginal vein was visualized. However short and long saphenous venous systems were normal. The patient was managed conservatively with compression stockings, limb elevation, appropriate skin care regimen and counseling.

DISCUSSION:

The lateral marginal vein or the vein of Servelle is the

embryonic persistence of a part of the superficial venous system, which usually occurs in association with Klippel Trenaunay Syndrome. When the venous trunk fails to mature during the embryonic period, it remains as the lateral marginal vein. This lateral marginal vein most commonly occurs along with lymphatic malformation. The veins here carry an extremely higher risk for DVT and pulmonary embolism owing to the lack of valves and the weakness of smooth muscle of the tunica media of the vessel wall.

Clinical features include the hallmark triad of capillary malformations that present as port wine stains on the affected limb, venous malformations in the form of varicosities along with limb hypertrophy that presents as asymmetrical limb growth impacting the mobility and thus the quality of life. Usual imaging studies include Doppler ultrasonography, MRI and CT angiography. Genetic testing, though not of good diagnostic value, may however be considered in atypical cases or for familial screening.

DIFFERENTIAL DIAGNOSES:

A special mention to Parkes-Weber syndrome characterized by multiple arteriovenous fistula causing venous hypertension, ulceration and high output cardiac failure. Other differential diagnoses include CLOVES (congenital lipomatous overgrowth vascular malformation with epidermal nevus and skeletal abnormalities) and MCAP syndrome (megalencephaly-capillary malformation), DCMO (diffuse capillary malformation with overgrowth), FAO (Fibro adipose overgrowth), Beckwith-Wiedemann syndrome characterized by hemihypertrophy, neonatal hypoglycemia, omphalocele, ear abnormalities, Maffucci syndrome-multiple enchondromas and plexiform neurofibromatosis in NF-1.[1]

MANAGEMENT:

KTS requires a multidisciplinary management that is tailored according to individual patient needs. Pain management and physiotherapy play a vital role. Rapamycin is newer therapy that works by inhibiting PI3K/AKT/mTOR pathway and can stop the progress of vascular malformations. [1] Pregnant patients may require antithrombotic therapy for DVT prophylaxis [5]. Low molecular weight heparin is used to reduce the risk of intravascular coagulation in patients undergoing surgical procedures. Surgical management for Klippel Trenaunay is still debatable as the recurrence rate is very high.

Debulking procedures have limited use and may damage venous as well as lymphatic structures leading to increased edema in the affected limb. Recent advances in Interventional procedures for KTS include sclerotherapy, radiofrequency ablation, endovenous laser ablation [9]. Radiofrequency ablation is gaining popularity because of great patient satisfaction owing to reduced pain and complications. Endovenous ablation is contraindicated in some literature because of a higher risk of VTE in an already prone to DVT patient [1]. Flebogrif system(mechanochemical ablation) has been found to have a very high success rate in treating chronic venous insufficiency in patients with KTS. Furthermore, Flebogrif is minimally invasive, has a shorter learning curve and allows faster resumption of physical activity.[8]

COMPLICATIONS:

Cellulitis of the affected limb can lead to sepsis and septic shock. Venous thromboembolism and subsequent pulmonary embolism are some of the dreaded complications of Klippel Trenaunay syndrome. Further complications may include gastrointestinal vascular malformations causing chronic occult blood loss requiring multiple transfusions, life-threatening bleeding associated with disseminated intravascular coagulopathy. [1][3] The patient may develop pulmonary hypertension in the long run owing to recurrent pulmonary embolism secondary to deep venous thrombosis.

CONCLUSION:

Patient education and awareness is extremely important in cases of Klippel Trenaunay syndrome. The primary caregiver plays a crucial role in coordinating amongst various specialists to improve the outcomes in the long run. A dermatologist may help in confirming the diagnosis and planning laser therapy for port wine stains. An orthopedic expertise may help with limb length discrepancy issues for epiphysiodesis and debulking procedures. Vascular surgeons may be required for sclerotherapy and open surgical procedures. Appropriate skin care, close follow up and timely multidisciplinary treatment should prevent life threatening complications and ensure that Klippel Trenaunay syndrome, though a progressive multisystem disease, remains manageable.

CONFLICT OF INTEREST: None

INFORMED CONSENT: Informed consent was obtained for publication of case report and clinical photographs of the patient.

REFERENCES:

1. Srividya Nagannathan, Prasanna Tadi ; Klippel Trenaunay syndrome https://www.ncbi.nlm.nih.gov/books/NBK558989/
2. Chagas CAA, Pires LAS, Babinski MA, Leite TFO. Klippel-Trenaunay and Parkes-Weber syndromes: two case reports. J Vasc Bras. 2017 Oct-Dec;16(4):320-324. [PMC free article] [PubMed]
3. Sharma D, Lamba S, Pandita A, Shastri S. Klippel-trénaunay syndrome - a very rare and interesting syndrome. Clin Med Insights Circ Respir Pulm Med. 2015;9:1-4. [PMC free article] [PubMed]
4. Vahidnezhad H, Youssefian L, Uitto J. Klippel-Trenaunay syndrome belongs to the PIK3CA-related overgrowth spectrum (PROS). Exp Dermatol. 2016 Jan;25(1):17-9.[PubMed]
5. Martinez-Lopez A, Salvador-Rodriguez L, Montero-Vilchez T, Molina-Leyva A, Tercedor-Sanchez J, Arias-Santiago S. Vascular malformations syndromes: an update. Curr Opin Pediatr. 2019 Dec;31(6):747-753. [PubMed]
6. Alwalid O, Makamure J, Cheng QG, Wu WJ, Yang C, Samran E, Han P, Liang HM. Radiological Aspect of Klippel-Trénaunay Syndrome: A Case Series With Review of Literature. Curr Med Sci. 2018 Oct;38(5):925-931. [PubMed]
7. Sung HM, Chung HY, Lee SJ, Lee JM, Huh S, Lee JW, Choi KY, Yang JD, Cho BC. Clinical Experience of the Klippel-Trenaunay Syndrome. Arch Plast Surg. 2015 Sep;42(5):552-8.[PMC free article] [PubMed]
8. https://www.mdpi.com/2079-4991/11/2/544/review_report
9. https://www.ejves.com/article/S1078-5884(19)31237-7/fulltext Endovascular treatment of klippel -Trenaunay syndrome- European journal of vascular and endovascular surgery.