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# Klippel Trenaunay syndrome : a rare case presentation involving multiple sites

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## Abstract

Klippel-Trenaunay syndrome (KTS) is a rare congenital disease, characterized by a triad of clinical features: (1) capillary malformations, manifesting as a “port wine stain”, (2) vascular anomalies, mostly varicose veins and (3) bone and/or soft tissue hypertrophy, usually of one lower extremity. The symptoms are frequently accompanied by lymphatic abnormalities that in some cases may lead to lymphedema. KTS is mostly benign in the course. Nevertheless, patients with KTS are at higher risk of developing deep vein thrombosis (DVT), pulmonary thromboembolism. Recurrent episodes of thrombophlebitis, dermatolymphangitis or internal bleeding.The diagnosis are from clinical presentations. Management in KTS should be individualized, minimally invasive and involve multidisciplinary care of the patient.

**Keywords:** Klippel-Trenaunay syndrome, vascular malformation, DVT, lymphedema, embolism

## Introduction

Klippel-Trenaunay Syndrome (KTS) is a rare congenital disorder[1]with an incidence of 3-5/100,000, characterised by capillary, venous malformations and soft tissue or bone hypertrophy with overgrowth of the affected extremity. Its aetiology remains unknown. Klippel-Trenaunay syndrome affects both genders equally. The cutaneous capillary malformation presenting as a port-wine stain and limb hypertrophy are usually noted at birth. Some cases presenting with atrophy and reduced growth of the affected limb have been described. Venous malformations may be present at birth or appear during infancy. The lower extremity is the most commonly affected. However, it can involve upper limbs and extends to the trunk. Doppler ultrasound allows identifying abnormalities of the venous system and MRI helps to characterise vascular malformations. Complications may include limb-length discrepancy leading to impaired gait and pain, thromboembolism, bleeding, venous insufficiency and soft-tissue infection.[2-4]

Diagnosis is mainly by clinical. Classical clinical triad, is enough for the diagnosis of KTS. Patients with at least two of the three features have been classified as having an incomplete form of KT syndrome. Non invasive radiological studies like radiographs of the involved limbs, Doppler study, MRI is helpful for confirmation of diagnosis[4].

Conservative treatment is the main stay for this condition. It involves multidisciplinary approach involves paediatrician, vascular surgeons, orthopaedic surgeons, phlebotomists etc. Symptomatic cases

accounts for interventions like minimal invasive surgeries i.e. pulse- dye laser therapy for port wine stain, Serial sclerotherapy with absolute alcohol or sodium tetra decylsulphate, polydocanol; or complete excision of AV malformations.[5]

**Case Report**

17 years old male patient presented with c/o left leg pain, multiple swellings over left leg(figure-1), right chest(figure-2), right thigh(figure-3) and lower angle of right scapula(figure-4). The swellings were soft, nontender, globular and hyper pigmented. No discharge from the swellings. The local temperature was normal, no visible or palpable pulsations.

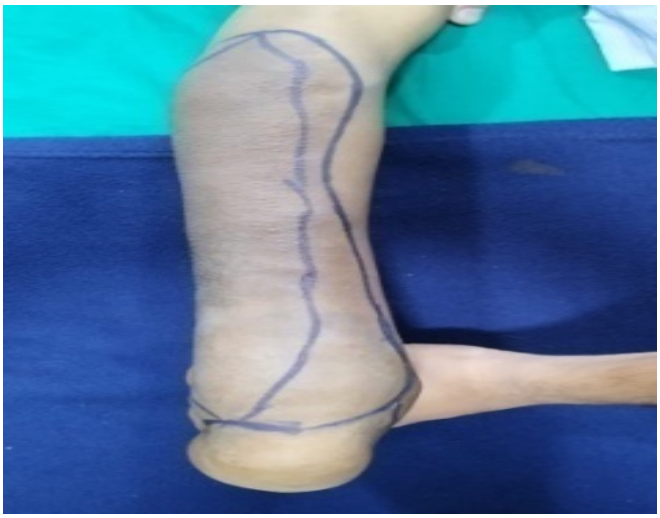

Figure 1

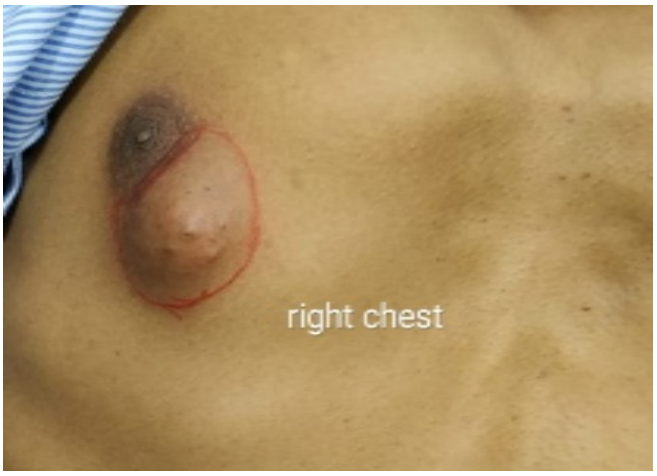


Figure 2

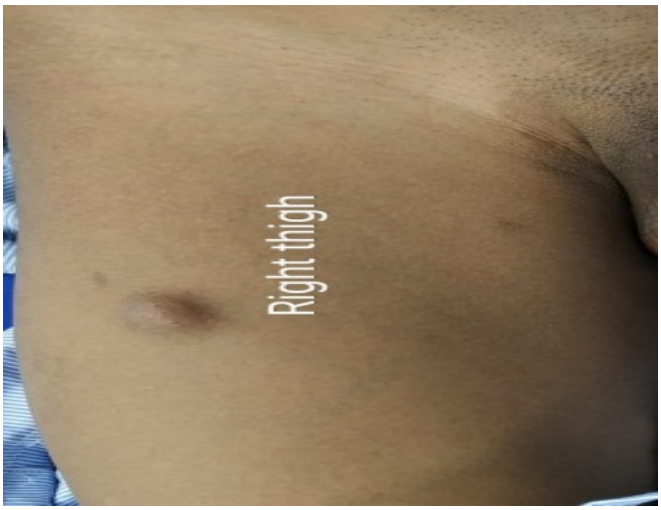


Figure 3

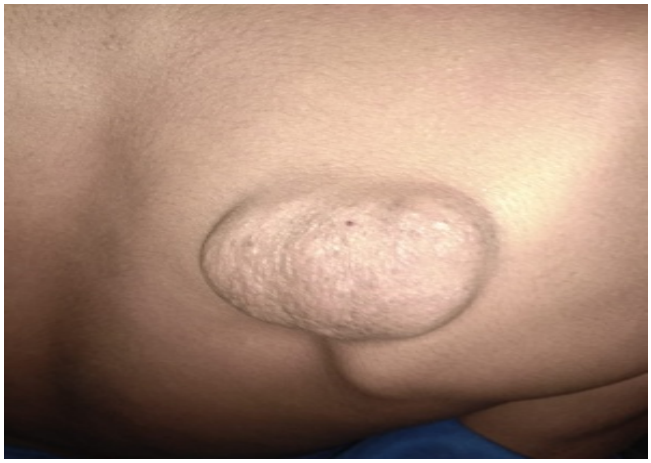

Figure 4

Doppler study(Figure-5) of left lower limb vessels shows popliteal vein, posterior tibial vein & anterior tibial vein appears normal but short saphenous vein is dilated tortuous having varicosity with echogenic content. Multiple dilated tubular superficial vascular channels found confirming A-V malformation. Deep veins appear normal in caliber and blood flow. The CT angiography (Figure-6 & 7) shows bunch of vessels in the posterior aspect of left leg s/o A-V malformations. MRI left leg showed bony hypertrophy with excessive soft tissue shadow around lower end tibia-fibula.

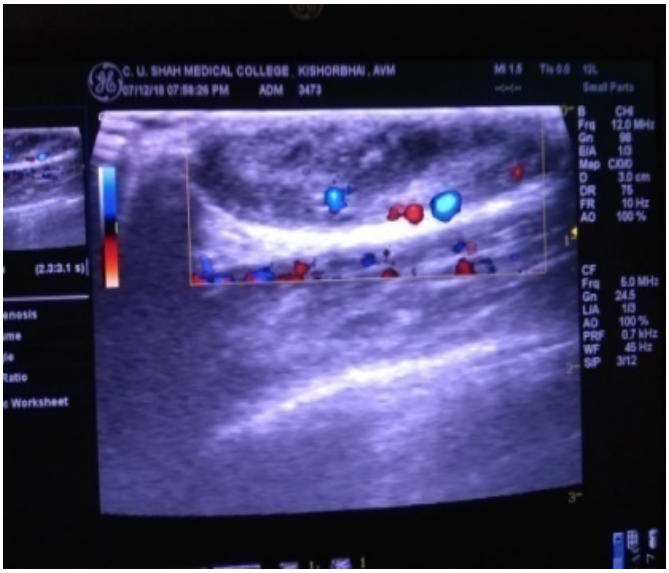


Figure 5

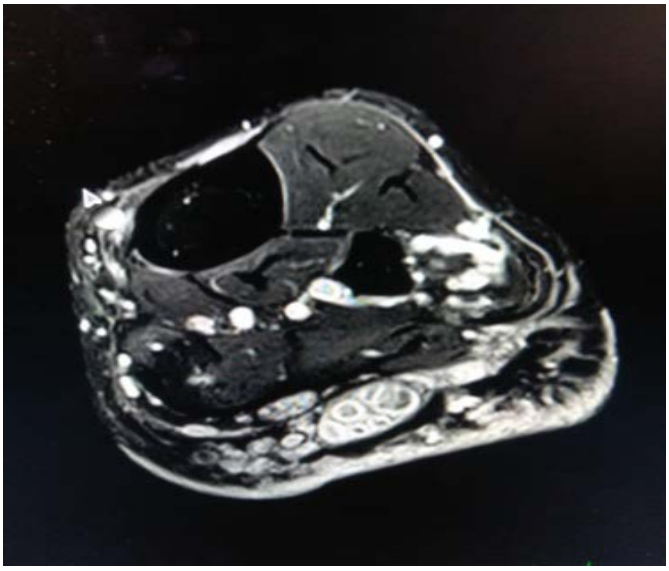

Figure 6

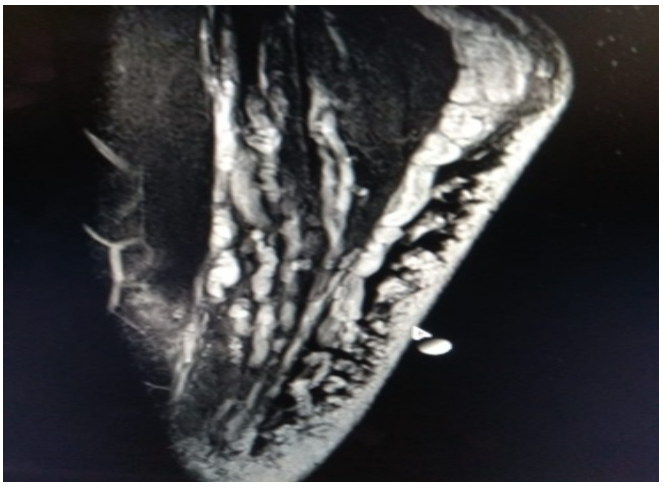

Figure 7

**Surgical Technique**

The procedure was done under general anesthesia, in supine and prone position. All malformations were marked prior to incisions. The incision taken, beginning from the ankle to the knee joint, over the leg malformation. Skin flaps raised with adequate thickness. The vascular malformation and hypertrophied connective tissue completely excised including deep fascia. (Figure-8 & 9) The closure of skin done, after adequate local hemostasis. Single vacuum drainage tube inserted. Remaining A-V malformation over thigh, periareolar and infrascapular area removed en toto and skin approximated. Excised specimen sent to histopathological examination (figure-10).

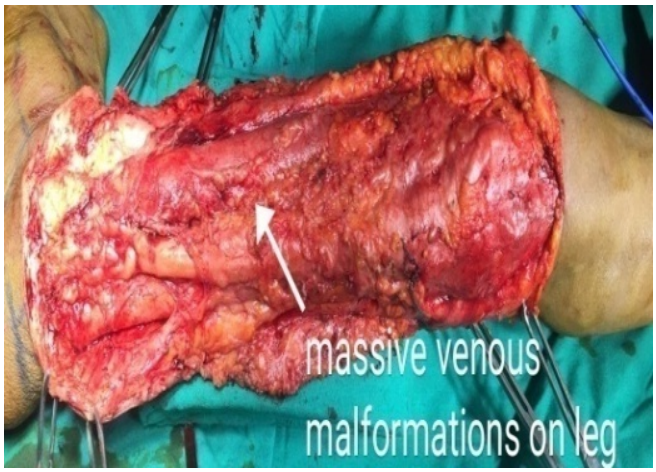


Figure 8

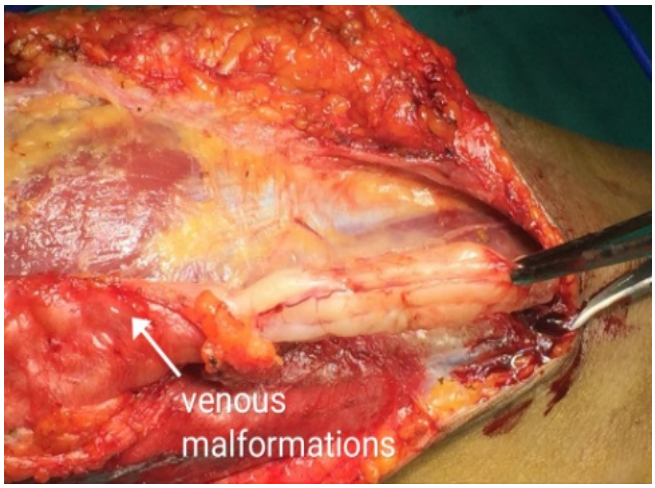


Figure 9

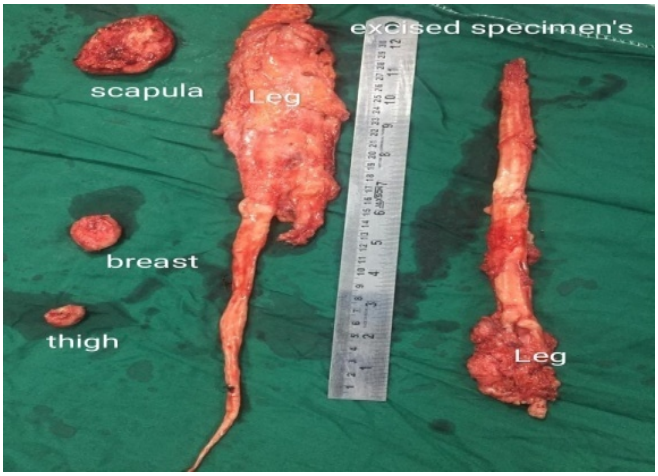

Figure- 10

**Discussion**

Two French physicians Maurice Klippel and Paul Trenaunay described two patients with haemangiomatous lesions of the skin associated with asymmetric soft tissue and bone hypertrophy, and coined the term "naevus variqueux osteohypertrophique".[6]This rare syndrome characterized by clinical triad of (1) capillary malformation (port-wine stain); (2) soft tissue and bony hypertrophy; and (3) atypical mostly lateral varicosity, was later termed as Klippel Trenaunay Syndrome (KTS). KTS should be distinguished from Parkes-Weber syndrome, a mixed, high-flow, high shunt arterio-venous malformation, since clinical features, management and prognosis of these two entities are distinctly different.[7] KTS has a wide spectrum of presentation, from truncular to extratruncular, from infiltrating to limited forms, containing primarily three anomalous vascular elements: veins, capillaries and lymphatics.

The aetiology of KTS is unknown. KTS is most commonly a sporadic event. Several theories have been proposed, including (1) Servelle's theory of a primary obstruction of the venous system resulting in venous hypertension and therefore development of abnormal venous pathways and tissue overgrowth; (2) failure of regression of the lateral limb bud vein; and (3) alteration of the tight balance between angiogenesis and vasculogenesis, which is controlled by numerous genes, among other theories. Berry *et al* in 1998 speculated that in KTS there is an alteration in vascular remodelling, perhaps at the level of altered angiopoietin-2 antagonism. The various case reports of KTS are present in world literatures, still incidence and genetic predisposition of this rare disease has not yet established.[8]

Classic clinical triad includes capillary malformations (port wine stain), a usually longer and larger extremity because of soft tissue and bone hypertrophy, and atypical mostly lateral superficial varicosity. Deep vein anomalies like venous hyperplasia to frank aneurysm, valve hyperplasia have been described. The venous malformations frequently present as persistence of embryonic veins, of which the lateral marginal vein (the vein of Servelle) has been the most typical finding found in 68-80% of patients. This vein originates from the lateral aspect of the foot and courses upwards along the lateral border of the leg. The vein is usually thick walled and strong, it is located immediately under the skin and it is incompetent along its entire length due to the absence of venous valves. The bony abnormalities may affect all bones in an extremity or limited to one or two bones. Single limb involvement is found in 80-85% patients.[9]

The absolute indications of treatment are haemorrhage, infections, acute thromboembolism or refractory ulcers. The management of KTS has been largely conservative. The compression therapy has been the mainstay of conservative treatment in the form of an elastic garment or compression bandage. This has been beneficial in managing both lymphedema and chronic venous insufficiency. Local wound care, compression

dressings, special orthopaedic footwear and lifestyle modification may also be required to manage activities of daily living and improve the function of the limb.[10] The cases with normal deep veins, complete surgical resection of the marginal vein is the best form of treatment. This vein may have very large perforators to the deep veins, so adequate exposure of the veins is recommended, especially for large perforators to be ligated. If deep veins are hypoplastic, marginal vein can be resected because deep veins are able to dilate spontaneously to almost normal size after resection.[10] The aplastic deep vein is absolute contraindication, for resection of marginal vein. The use of subfascial endoscopic perforator surgery (SEPS) in patients with large incompetent perforating veins and venous ulcers has been useful and some patients benefit from deep venous reconstructions.[11]

Our case was the symptomatic one. Swelling was painful and bad cosmetic appearance was an issue for the patient, so we went for operative management for this patient. We excised all the swellings en toto and made the patient symptom free. Patient was fully ambulatory and painless postoperatively.

**Conclusion**

The management of patients with K-T syndrome continues to be primarily nonsurgical, but those patients with patent deep veins can be considered for excision of symptomatic varicose veins and hypertrophied connective. This disease has a high recurrence rate, but clinical improvement is significant and reoperations can be performed if needed. Diagnosis is purely clinical. There is no cure for this disorder, majority cases management is conservative with lifelong follow up. The approach should be multidisciplinary as K-T syndrome affects multiple systems. We conclude that the management of KTW syndrome includes careful and early diagnosis. Complete excision possible with acceptable cosmetic look and quality of life. .

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