

Klippel Trenaunay Syndrome: A Rare Vascular Disorder

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ABSTRACT

Klippel-Trenaunay-syndrome (KTS) is a rare vascular disorder consisting of a triad of features including capillary malformations, varicose veins and soft tissue abnormalities. It overlaps with few other rare vascular disorders such as ‘Sturge Weber syndrome’ and ‘Parks Weber

syndrome’ with few notable distinguishing features. The diagnosis is clinical, however, imaging is necessary for confirmation. The treatment is conservative and individualized. In this case report, we present a variant of KTS which demonstrates only two of the three features.

Keywords: Klippel Trenaunay Syndrome; Capillary Malformations; Varicose Veins; Sturge Weber Syndrome; Parks Weber Syndrome

CASE REPORT

A 25 years old male patient, a mechanic by profession, presented in the surgical outpatient department with complaints of prominent veins on both lower limbs for the last 12 years. Veins were small and progressively increased in size for last one year. Veins would empty on lying down and would be prominent on standing. For the last one year, there was continuous itching and mild pain in lower limbs on walking and standing. There was no history of trauma, insect bite, bleeding disorder, fever or abdominal pain. The patient had a history of pulmonary tuberculosis (TB) three years ago and he had taken anti-tuberculous therapy (ATT) for 6 months. His mother died of TB and father had decompensated liver disease secondary to hepatitis C virus infection. He was addicted to tobacco chewing and occasionally drank alcohol. On examination, there were bilateral tortuous prominent veins from the ankle up to the thigh, more marked on the lateral aspect of limb but not in the territory of the long saphenous vein (Figure 1). Veins were compressible, non-tender with negative Morrissey’s cough impulse. There was slight pigmentation on the left foot and ankle with patchy areas of lipodermatosclerosis. These veins became less prominent on lying down. Tourniquet test revealed bilateral competent saphenofemoral junctions. There was a hard bony outgrowth on the medial aspect of the left knee

and the 2nd toe of the right foot was longer than the left (Figure 1 and 2). Both lower limbs were grossly enlarged in cross-sectional diameter. There was no excessive sweating in this patient. There was no capillary malformation (port wine stain) noted in this patient. Ultrasound duplex scan revealed abnormal dilated superficial veins with normal deep veins and competent sapheno-femoral and sapheno-popliteal junctions. There was no bony outgrowth from bone but a calcified varix noted on X-ray knee joint (Figure 3). Further confirmation was achieved with Magnetic Resonance angiography. Initial impression of Klippel-Trenaunay-syndrome (KTS) was made and discussed with the vascular surgeon. This patient was been discharged with above knee stockings and with regular follow up.

DISCUSSION

Klippel-Trenaunay-syndrome (KTS), a rare vascular malformation disorder, can present in many ways from simple mild forms of port wine stains and few varicose veins of cosmetic concern to severe disability associated with limb over-growth and large abnormal varicose veins [1]. An extreme form of the disorder may lead to recurrent rectal bleeding, chronic pain, thromboembolism, skin infections, and arthritis. KTS has been classically described as a triad of capillary malformation, atypical large abnormal varicose veins and soft tissue overgrowth of

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Figure 1: Bilateral tortuous prominent veins from the ankle up to the thigh, more marked on the lateral aspect of limb but not in the territory of the long saphenous vein



Figure 2: Hard bony outgrowth on the medial aspect of the left knee



Figure 3: There was no bony outgrowth from bone but a calcified varix noted on X-ray knee joint



extremity [2]. Presence of any two of the three features of the triad in a patient may be labeled as an atypical or incomplete form of KTS [3].

The port wine stain, a cutaneous malformation often included in KTS, is dermatomal in distribution and may or may not blanch with pressure [4]. It is usually red to purple in color. Classically, it is found on the same extremity of the limb from buttocks to thigh but in rare variants, it may be found bilaterally, in contralateral limb, on the face, trunk, neck or limited to the midline of body [5].

Unlike more common varicosities which are found on the medial aspect of the lower extremity, KTS varicosities are very large, abnormal and distributed laterally from foot to groin region [6]. These varicosities may extend above the groin region into the pelvis and visceral organs such as the rectum or urinary bladder and may cause troublesome recurrent rectal bleeding and hematuria respectively [7]. Cellulitis, dermatitis, ulceration, thromboembolism, or hemorrhage may also occur with these varicosities.

Bone and soft tissue hypertrophy are present in the majority of KTS cases [8]. The limb discrepancy is secondary to soft tissue and bony growth both in length and circumference. Mostly, only one limb is affected but the combination of upper and lower extremities have also been reported.

There are also some other rare vascular disorders that may overlap with KTS such as, Sturge Weber syndrome and Parks Weber syndrome. There are few differences that help to distinguish among these disorders. In Sturge Weber

syndrome, capillary malformation (port wine stain) occurs on face and eyes in contrast to extremities in KTS [9]. The port wine stain in Sturge Weber syndrome is cutaneous but sometimes it may be deep and involve leptomeninges and may cause neurological complications like hemiparesis, mental retardation and epilepsy. Similarly, Parks Weber syndrome also overlaps KTS but the most important difference is the presence of arterio-venous malformations in former [10].

Various theoretical explanations have been proposed regarding the pathophysiology of KTS yet the etiology remains an enigma. The management is multidisciplinary involving interventional radiologists, orthopedic and vascular surgeons. The usual treatment is conservative in most cases like above knee stockings, prevention from trauma, infection and laser ablation for small varicosities [11].

CONCLUSION

The diagnosis of KTS is purely clinical but one has to differentiate it from Parks Weber and Sturge Weber syndrome by the absence of arterio-venous malformations and the presence of port wine stain on face and neck respectively. The management is multidisciplinary and individualized with lifelong follow up.

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