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Case Report

Klippel-Trenaunay Weber Syndrome: A Rare Case Report

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ABSTRACT

Klippel-Trenaunay syndrome (KTS) is a rare congenital condition usually presenting with port wine stains, excessive growth of soft tissue and bones, and also varicose veins which most commonly occurs in the legs, but it also may affect the arms, face, head, or internal organs .This syndrome is has a huge individual variability, given that the majority of patients do not clearly present this classic trio. We are presenting a case of Klippel-Trenaunay Syndrome in a 28-year-old male patient presenting with varicosity in lateral aspect of right leg. He had many vascular abnormalities from birth which increased in time such that he had surgical interventions. On careful examination other components of the syndrome were found. The diagnosis was made with x-rays, HRCT chest and Doppler studies of the lower limb vessels. He is currently being managed conservatively with dressings, antibiotics and analgesics. Patient was followed up monthly for any abnormality.

Keywords: Hypertrophy; Port-wine stain; Vascular malformations; Klippel-Trenaunay Weber syndrome

INTRODUCTION

Klippel-Trenaunay Syndrome (KTS) is a rare congenital disorder [1] with an incidence of 3-5/100,000. It is characterized by a triad of vascular malformation of capillary, venous and lymphatic vessels (hemangioma or port-wine stain), venous varicosities mostly involving lateral venous system, and bony or, soft-tissue hypertrophy. The diagnosis of KTS can be made if any two of the three features are present. Capillary malformations may be absent in the atypical form. [2]

The difference between KTS and Klippel-Trenaunay-Weber syndrome (KTWS) is that the latter includes significant arteriovenous malformations in the affected extremity. [3]

The etiology is obscure; however, it has been suggested that this condition results from a paradominant mutation of the VG5Q gene, causing the abnormality of the mesoderma during fetal growth, making a secondary intra-uterine involvement possible. [4,5]

We are presenting a case of uncomplicated KTW syndrome and a brief review of the literature. Our patient presented with varicosity of veins in the surgical department. Patient was transferred to Medicine department for raised creatinine. Further on examination and history patient was found to have port-wine stain, repeated varicosities, for which he was operated earlier once. Patient was found to have vascular anomalies on HRCT chest and found on upper GI scopy. In rare cases, patient can present with cerebral malformations. Management of this syndrome should aim to correct any of the abnormalities present, if technically possible, and if the abnormality is causing symptoms.

CASE REPORT

A 34-year-old male patient presented to the surgery department of Dr. D Y Patil Medical College and Research centre, Kolhapur in the month of April, 2018 with varicose veins. Patient was transferred to Medicine department for raised serum creatinine. Patient gave complaints of dilated vein over outer aspect of left lower limb from a long time. He reported that the dilatation over the limb increased during walking and standing. It subsided when lying down with the limb raised. Cutaneous pigmentation was present over the left foot. Patient was operated for right varicosities 3 years back. Later on he developed varicosities of left lower limb. There was history of ulceration over left foot, difficulty in walking, pain in legs. There was no history of paresthesia, or cellulitis.

Patient also gives history of episodes of haemoptysis, haematemesis, melena and hematochezia occasionally.

On examination multiple varicose veins were present over the lateral aspect of left lower limb extending from above the ankle to the upper thigh (Fig 1). Brodie-Trendelenburg test showed sapheno-femoral valve to be incompetent.

Perthes test demonstrated competent deep venous system.

Port-wine stain over anterior aspect of foot.

All routine investigation done was normal except for mild anaemia. Repeat serum creatinine was within normal limits.

Colour Doppler of the patient revealed multiple varicosities all over the leg & lower thigh region, few showing thrombus formation within competent deep venous system. Mid thigh & medial leg perforators could be visualized & found normal in size & flow. Sapheno-femoral & sapheno-popliteal junctions were patent. Upper gastro-intestinal endoscopy showed few Grade 1 oesophageal varices. HRCT chest showed vascular pooling of blood in the infraclavicular region.

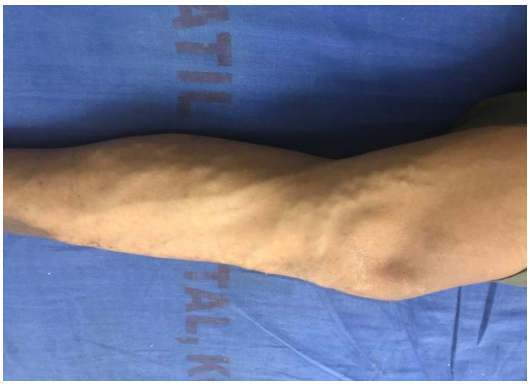


Figure 1: Leg affected by Varicose veins

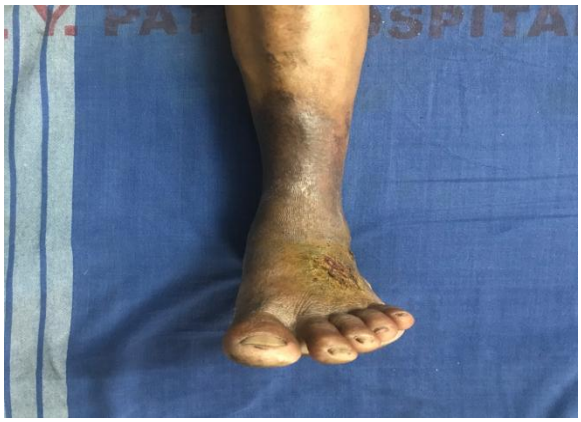
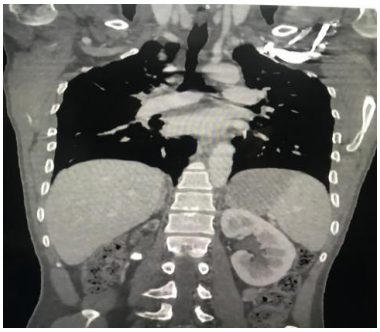
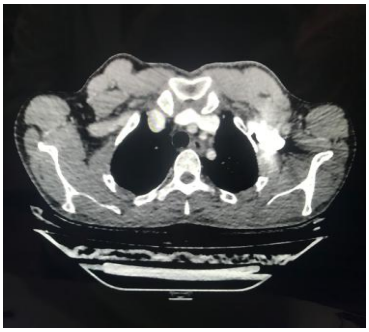


Figure 2: Leg showing prominent port-wine stain with lipodermatosclerosis.





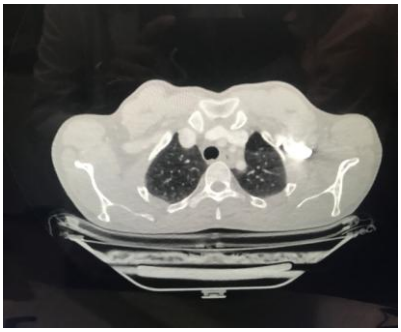


Figure 3: High Resolution CT scan of Lungs
Figure 4 and 5: Irregular pooling of contrast seen in the anterior chest wall in left infra clavicular region- Vascular dysplasia as seen in KT Syndrome.

DISCUSSION

About a century ago, 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". [7] This rare syndrome characterized by clinical triad of (a) capillary malformation (port-wine stain); (b) soft tissue and bony hypertrophy; and (c) 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 arteriovenous malformation, [8] since clinical features, management and prognosis of these two entities are distinctly different. 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.

A series of 252 patients with KTS was studied at Mayo Clinic, Rochester between January 1956 and January 1995. It showed presence of capillary malformations (port-wine stains) in 246 patients (98%), varicosities or venous malformations in 182 (72%), and limb hypertrophy in 170 (67%). All three features of KTS were present in 159 patients (63%), and 93 (37%) had two of the three features.

The aetiology of KTS is unknown. KTS is most commonly a sporadic event. Several theories have been proposed, including (a) 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; (b) failure of regression of the lateral limb bud vein; and (c) alteration of the tight balance between angiogenesis and vasculogenesis, which is controlled by numerous genes, among other theories. [9] 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. [10] Various case reports of KTS are present in world literatures. Still, incidence and genetic predisposition of this rare disease has not been established.

Imaging has an important role in the diagnosis and evaluation of KTS. At plain radiography, phleboliths in a young patient are usually diagnostic for venous malformations and are manifestations of prior hemorrhage or thrombus. Barium studies can show luminal narrowing of the affected small and large bowel that is distensible, with a scalloped mucosal outline caused by the presence of varicosities or submucosal vascular malformations. [7,12] Sonography may be used to identify the abnormal veins and varicosities. Computed tomography of the abdomen and pelvis provides a simple, noninvasive means of assessing visceral vascular malformations. [13] MRI is performed to assess the soft-tissue extent of vascular malformations in patients with KTS. [13] The role of MR angiography in analyzing vascular malformations in KTS has not been well defined, but the modality has the potential to depict these lesions with better accuracy. [12] In cases of hemorrhage that require surgical intervention, preoperative angiography is done to check the anatomical structure involved and involvement of intestine, hence, guiding surgical resection. [12]

The absolute indications of treatment are haemorrhage, infections, acute thromboembolism or refractory ulcers. The management of KTS has been largely conservative. 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 lymphoedema 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. [11]

Complications are related to the vascular pathologic process. Complications include stasis dermatitis; thrombophlebitis;

cellulitis; with serious complications including thrombosis, coagulopathy, pulmonary embolism, congestive heart failure (in patients with arteriovenous malformations), and bleeding from abnormal vessels in the gut, kidney, and genitalia. [5,6,9]

KTS should be suspected in all infants with capillary malformations involving one extremity of the body from birth. Differential diagnosis for KTS is KTWS, Proteus Syndrome, Maffucci Syndrome, among other nonsyndromic capillary malformations of the skin. [12]

CONCLUSION

The management of patients with KTW syndrome continues to be primarily nonsurgical, but those patients with patent deep veins can be considered for excision of symptomatic varicose veins. 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 Management is conservative with lifelong follow up. The approach should be multidisciplinary as KTW affects multiple systems.

We conclude that the management of KTW syndrome includes careful and early diagnosis, prevention of the disease and complications and further treatment of complications if occurred.

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