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A Series of Cases of Klippel Trenaunay Syndrome with Unusual Presentations

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Abstract: *Klippel trenaunay syndrome (KTS) is characterized by triad of portwine stain, venous varicosities and_bone and soft tissue hypertrophy. It is not commonly seen in dermatological practise. Hypertrophy and vascular malformations are usually confined to single limbs. We report a series of 2 cases with different presentations and they attended to our OPD in a span of 6 months from June 2018 to December 2018. Management is usually conservative and surgical management for varicosities with lifelong follow-up.*
Keywords: *Klippel Trenaunay syndrome, limb hypertrophy, lymphangioma, port wine stain, syndactyly, varicosities.*

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I. Introduction

Klippel Trenaunay syndrome (KTS) is a rare congenital disorder with an incidence of 3-5/ 100000. It is characterized by triad of vascular malformations (port wine stain), venous varicosities and soft tissue or bone hypertrophy. It can be associated with lymphatic obstruction, lipodermatosclerosis, affection of abdominal and pelvic vasculature leading to various degrees of vascular malformations involving the gastro intestinal system, spleen, genitourinary and central nervous system. Usually present at birth or childhood.

II. Case reports

2.1. Case report 1:

16year old male born out of non consanguineous marriage, presented with bleeding per rectum since 3 months. History of similar complaints since 5 years occasionally. Associated with history of progressive enlargement of right lower limb since 11years. History of inability to walk without aid and associated with dragging pain on long standing since 2 years.

On examination he had hypertrophy of left lower limb and hypertrophy of right leg and foot. Portwine stains were present on both the lower limbs(Fig 1). On right side the portwine stain extended from lower abdomen to the thigh and leg. Angiokeratomas present over the left thigh. Multiple lymphangiomas were present over the right axilla, trunk and lower limbs (Fig 2). Bilateral feet had syndactyly and enlarged digits on right hand (Fig 5,6). Associated with edema of scrotum (Fig 4). Linear verrucous epidermal nevus present (Fig 3). There is marked limb girth and limb length discrepancy.

X-ray of both feet and right hand showed abnormal phalanges and metatarsals. Colour Doppler showed abnormal arteriovenous malformations on left side. Ultra sound abdomen was normal.

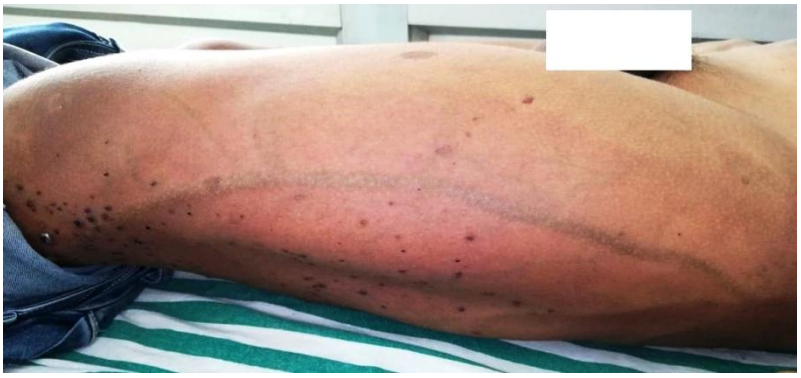

Figure 1: Large portwine stain

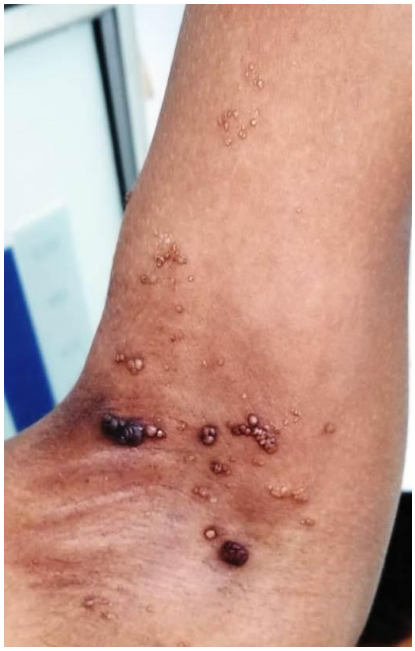

Figure 2: Lymphangiomas

Figure 3: Verrucous epidermal nevus

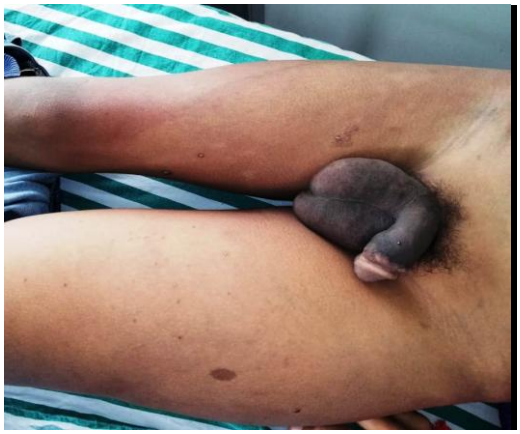

Figure 4: Limb girth discrepancy and scrotal edema

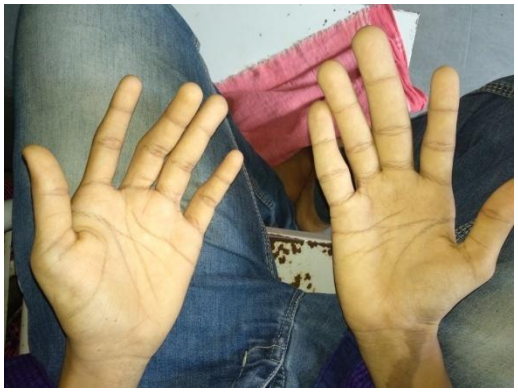

Figure 5: Enlarged digits of right hand

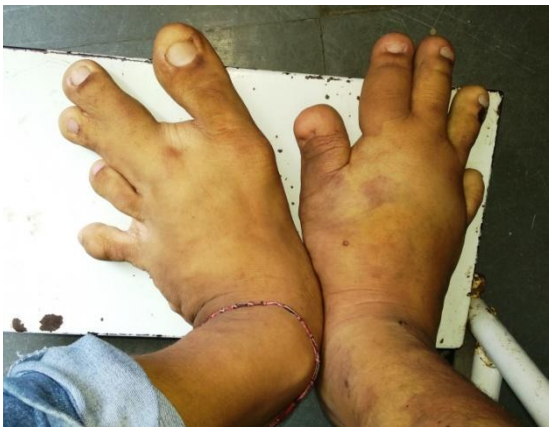

Figure 6: Syndactyly of bilateral feet

2.2. Case report 2:

18year old female born out of non consanguineous marriage presented with pigmented lesions over the left lower limb with history of occasional bleeding with minor trauma since 10 years. History of frequent ulceration over left leg and pain in left leg. No history of altered gait.

On examination multiple hyperpigmented verrucous plaques present over lower 1/3 rd of left leg and dorsum of left foot (Fig 7). Similar presentation in a case published by Sharma et al. Single hyperpigmented soft nodule 0.5× 0.6 cm present over left ankle. Limb length and limb girth discrepancy present but not marked(Fig 8) . Rise of temperature present over the entire left lower limb. Single soft swelling (? Varicosity) present over the junction of upper and middle 1/3rd of left leg on lateral aspect.
X- ray was normal. Colour Doppler revealed mild varicosities along short saphenous vein.

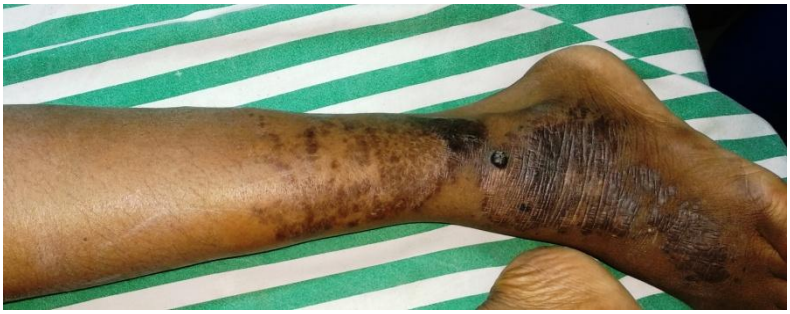

Figure 7: Hyperpigmented verrucous plaques

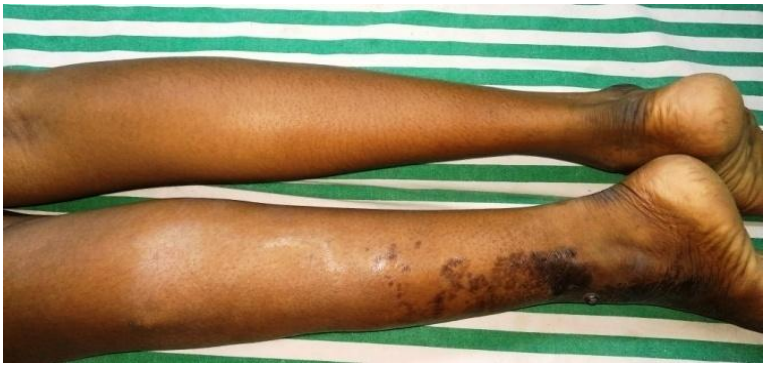

Figure 8 : Limb girth discrepancy

III. Discussion

KTS was described by two French physicians, Klippel and Trenaunay in 1900. It is a triad of capillary malformation, venous/lymphatic varicosity with soft tissue and bony hypertrophy. Some authors use the term KTWS (Klippel Trenaunay Weber Syndrome) to describe the conditions affecting those individuals who have significant arteriovenous malformations as one of the component of their KTS[4,6]. Other authors prefer to separate these two conditions and use the term Parkes –Weber Syndrome to describe the condition in those patients who have arteriovenous malformations in addition to KTS.[1]

It is now defined as a combination of capillary malformations, soft tissue, and bone hypertrophy, and lymphatic/venous malformations. The diagnosis of KTS can be made when any two of the three features are present.[1] Most cases of KTS are sporadic with both males and females equally affected, with no racial predilection, and usually manifests at birth, or during the childhood.[1]

Hypertrophy is the most variable of the three classic features of KTS. Enlargement of the extremity can be either bone elongation, circumferential soft-tissue hypertrophy, or both[6]. On clinical examination, hypertrophy often manifests as a leg-length discrepancy, although any limb may be affected. Significant limb-length discrepancy, defined as that amount that would necessitate orthopaedic intervention, is relatively uncommon, occurring in only 14% of patients in one study.[1,5]

Capillary malformations are the most common cutaneous manifestation of KTS.[4] Typically, capillary malformations involve the enlarged limb, although may be seen in any part of the body. The lower limb is the most common site of malformations found in approximately 95% of patients.[1] When found in the trunk, the malformations rarely cross the midline[5]. Other less common manifestations of KTS include thromboembolic episodes, thrombophlebitis, Kasabach-Merritt syndrome, hematuria, rectal or colonic bleeding, vaginal, vulval or penile bleeding in children with visceral and pelvic hemangiomas.[6]

The vascular changes found in KTS are congenital. Varicose veins are present in a majority of patients with KTS. Venous malformations can occur in both the superficial and deep venous systems.[1]

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.[8,9]

Vascular malformations that involved the gastrointestinal (GI) and genitourinary tracts have also been reported, and it can be a significant source of morbidity[1]. Patients having vascular malformations involving the bladder frequently usually have rectosigmoid or pelvic organ involvement. Rectal and bladder hemorrhage are few of the serious complications in pelvic vascular malformations and have been reported in 1% of cases[5]. Bleeding is usually the most common and earliest symptom reported in KTS patients with GI involvement. The most frequently involved sites of GI involvement include the distal colon and rectum.[1]

Genitourinary involvement in patients with KTS seems to occur in the more severe cases. The absence of limb varicosities or malformations does not exclude the presence of pelvic involvement. Gross hematuria, is generally recurrent and painless and is first clinical sign suggesting bladder involvement and frequently occurs early in life.[2,3]

Associated developmental defects usually seen are various and include polydactyly, oligodactyly, macrocephaly, blue nevi, pulmonary varicosities, cerebral aneurysm and pulmonary embolism[3].

Imaging has an important role in the diagnosis and evaluation of KTS. 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.MRI is performed to assess the soft-tissue extent of vascular malformations in patients with KTS.[1]

There is no cure for this disorder. Therapeutic objectives seek to improve the patient's condition and treat the consequences of severe lesions. Treatment of port-wine stains is usually done with pulsed dye laser therapy that yields better results when applied to lesions in the face and trunk, as compared to extremities. Nevertheless, it only contributes to the superficial treatment of hemangiomas. When varicose veins are present, compression stockings are recommended for venous insufficiency. Surgical treatment is only recommended in symptomatic cases of superficial varicose veins. The use of orthopedic braces is a good option to prevent the development of vertebral deformities in case of hypertrophy of the lower limbs. With time, corrective bone surgery may be necessary to treat significant limb length discrepancy, if present.[5]

Patients with KTS should be monitored at least annually and more frequently if clinical symptoms are present. If there is a progression of disease, imaging studies should be done, and proper intervention carried out if indicated.[1]

IV. Conclusion

KTS is a rare disorder and can involve multiple organs. Proper history should be taken to rule out systemic involvement. Hypertrophy of limbs may cause vertebral scoliosis and abnormal gait. Hence early diagnosis should be done to monitor progression and complications. Management is conservative with lifelong follow up.

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