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Dermatology

KLIPPEL-TRENAUNAY SYNDROME: A RARE CASE REPORT

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ABSTRACT Klippel-Trenaunay syndrome (KTS) is a rare syndrome characterized by triad of soft tissue and bony hypertrophy, capillary malformation (port-wine stain) of the skin and varicose veins. KTS can be diagnosed if any of the two features are present. We here report a fifty year old female with Klippel-Trenaunay syndrome.

KEYWORDS : Klippel-Trenaunay syndrome, port-wine stain, bony hypertrophy

INTRODUCTION:

Klippel-Trenaunay Syndrome is an infrequent disorder with three classical features namely hypertrophy more often of the lower limbs, with vascular anomalies and capillary malformations of the skin, coined the port wine stain [1]. It may be coupled with lymphatic obstruction, lipodermatosclerosis and vascular malformations of the central nervous system, gastro intestinal system and genito-urinary system [2, 3]. This condition frequently presents at birth with equal gender predilection [4].The various synonyms for Klippel-Trenaunay syndrome are Hemangiectasia hypertrophicans; Angio-osteohypertrophy; Nevus varicosus osteohypertrophicus syndrome; and Nevus verucosus hypertrophicans. When associated with arterio venous malformations termed as Klippel-Trenaunay-Weber syndrome. We report here a case of Klippel Trenaunay Syndrome in a fifty year old female.

CASE REPORT:

A fifty year old female presented to the skin OPD with complaints of erythematous patches over left thigh and leg for past 10 yrs, the patches were progressive in nature. Initially asymptomatic, but now for the past 1 year, patient had pain and swelling of the leg. General examination was normal. Dermatological examination revealed an ill defined erythematous patch over the entire left lower limb. The lesions were non blanching [Figure 1]. Limb girth was 2.5cm more over left thigh and leg compared to the right [Figure 2]. Varicose vein was not seen. No scoliosis and gait abnormality noted. Other skin, hair and mucosa were normal.

Skin biopsy revealed ectatic capillaries in the papillary and reticular dermis [Figure 3, 4]. Few scattered red blood cells were noted in ectatic capillaries [Figure 5].Skiagram of lower limb showed bony and soft tissue hypertrophy of the left limb [Figure 6, 7]. An arterial and venous ecodoppler study of the lower left limb was conducted and no anomalies were found. Antibiotics and pain relief were given. Patient was insisted on regular follow up.

DISCUSSION:

Klippel Trenaunay Syndrome is a rare congenital disorder in which blood vessels and/or lymphatic vessels fail to form properly [1]. The three key features that describe this condition are a port wine stain, caused by capillary malformations of the skin, vascular anomalies and hypertrophy of the soft tissues and bones [1, 4, and 5]. It was in the year 1900 two French physicians named Paul Trenaunay and Maurice Klippel described two patients with a triad of port wine stain, varicosities of an extremity and hypertrophy bones and soft tissues of the affected limb [3, 6]. Parks Weber few years later independently stated a syndrome with the above said triad and the presence of arterio-venous malformations [7,8].

The exact aetiology of KTS is unknown; however very few theories have been postulated. Popularly viewed theory is the one proposed by Baskerville et al [4] .It is a mesodermal defect encountered during embryogenesis resulting in maintenance of microscopic arteriovenous communications ending in KTS. Other theories are that it might occur due to a single gene defect (Happle, 1993). Or rarely could be inherited as an autosomal dominant trait (Cebllos-Quintal et al, 1996). Whelan et al. described a balanced 5:11 translocation (an equal exchange of material between two chromosomes with no genetic information lost or added). There is neither racial nor gender predilection [4,5].

The diagnosis is mainly clinical, imaging studies like Ultra sonogram, contrast enhanced MRI and Doppler study are required to find out the extent of lesion thereby helps to plan interventions if indicated. Treatment is indicated to reduce the symptoms and the risk of complications. Active interventions are needed only for localized lesion and presence of serious complications like bleeding or cardiac failure.

Currently, the vascular lesions are treated with flash lamp-pumped pulsed dye laser and it is most effective when performed early. The varicose veins can be corrected surgically by vein ligation, stripping, resection and amputation. Sclerotherapy can be done by chemicals like sodium tetradecyl sulfate, ethanolamine, and absolute ethyl alcohol. Debulking procedures are often not recommended as they damage venous and lymphatic structures resulting in increased edema of the affected limb. Chronic venous insufficiency, bleeding from capillary or venous malformations, lymphedema and recurrent cellulitis can be corrected by compression therapy. It also protects the limb from trauma.

Cellulitis and thrombophlebitis are treated with analgesics, corticosteroids, antibiotics, and elevation. Complications owing to hemangioma are secondary infection and bleeding [7]. Complications of varicose veins include dermatitis, cellulitis, leg ulcers, pulmonary embolism and thrombophlebitis. Scoliosis and gait abnormalities are sequelae to limb hypertrophy. Heel inserts are normally sufficient for 1.5 cm or less limb length discrepancy. If leg length discrepancies are more than 2.0 cm, it may be treated by epiphysiodesis in the growing child.

CONCLUSION:

Patients with KTS should be monitored at least annually to prevent complications. Clinical follow up is sufficient with stable disease. If the disease progresses, imaging studies might be required and medical or surgical intervention should be pursued if indicated.

ACKNOWLEDGEMENT
None

CONFLICT OF INTEREST
The authors declare that they have no conflict of interest.

LEGENDS TO FIGURE:
FIGURE 1: Clinical photograph showing non blanching port wine stain on the left lower limb.
FIGURE 2: Clinical photograph showing left lower limb hypertrophy.
FIGURE 3: Photomicrograph showing ectatic capillaries.
FIGURE 4: Low power view (10 X) showing ectatic capillaries in the papillary and reticular dermis.
FIGURE 5: High power view (40 X) showing few scattered red blood cells in the ectatic capillaries.
FIGURE 6, 7: Skiagram showing bony and soft tissue hypertrophy of left limb

FIGURE 1:

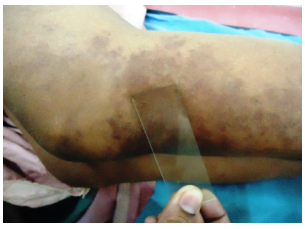


FIGURE 2:

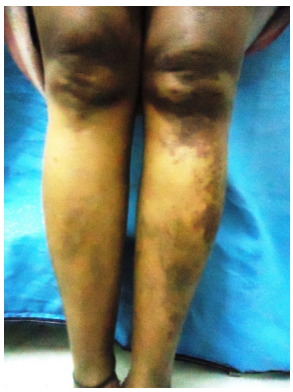


FIGURE 3:

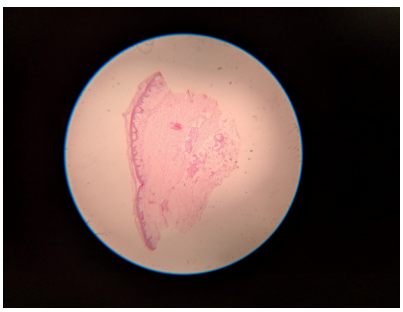
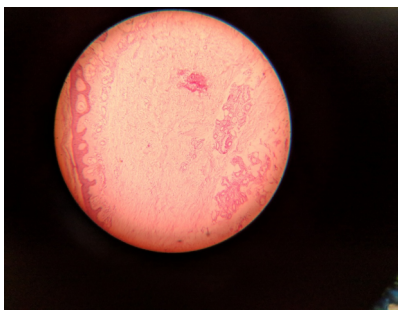


FIGURE 4:



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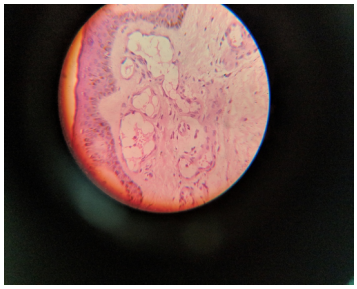

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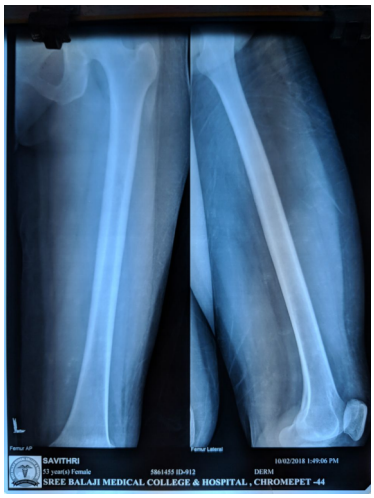
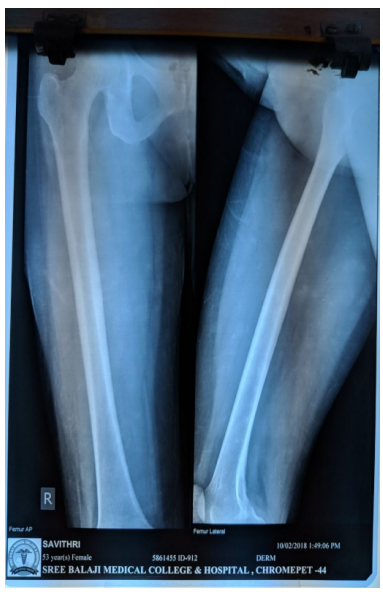


FIGURE 7:



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