Case report:

Klippel Trenaunay Syndrome - A case report

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Abstract:
Klippel-Trenaunay Syndrome (KTS) is a sporadic disorder characterized by the triad of vascular malformation (capillary haemangioma or port wine stain), venous varicosity and soft tissue and/or bony hypertrophy. We report here a case of Klippel-Trenaunay syndrome with review of literature.

Key Words: Klippel-Trenaunay Syndrome, Port wine stain, Venous varicosity

Introduction:
Klippel-Trenaunay syndrome (KTS) is a rare disorder with an incidence of 3-5/1, 00,000.¹ It is characterized by the triad of vascular malformation (capillary haemangioma or port wine stain), venous varicosity and soft tissue and/or bony hypertrophy. The vascular malformation is usually limited to a single extremity, though multiple extremities can be involved. Alternative names given for Klippel-Trenaunay Syndrome are Klippel-Trenaunay Weber syndrome; Angio osteo hypertrophy; Nevus varicosus osteohypertrophicus syndrome; Hemangiectasia hypertrophicans and Nevus verucosus hypertrophicans

Case report:
A seven year old female child was brought to our OPD by her mother with complaints of enlarged left lower limb and left size of face gradually since birth (Figure 1 & 2). Detailed history revealed that patient was born out of a non consanguineous marriage with normal perinatal history. After birth, mother noticed some spots on the lower back and left foot of the patient along with gradual enlargement of the left leg. Clinical examination showed full, range of motion at all joints of left lower limb with soft tissue oedema with no hypertrophic changes in left upper limb. Muscle power and sensations were normal and she had normal pulsations in left lower limb. She had a large port wine stain on the dorsum of left foot and lumbo sacral area also (Figure 3 & 4). Clinically left lower limb showed hypertrophic changes as compared to right side and left great toe and second toe was hypertrophied. There was hemi facial hypertrophy on left side. Patient also had improper dentition (Figure 5).

Patient was investigated with X-rays of bilateral lower limb with pelvis which did not show any bony hypertrophic changes (Figure 6). Ultrasonography of abdomen and pelvis was unremarkable. Doppler studies of left lower limb showed venous incompetence with venous malformation in the gluteal region and left thigh leg and foot with heterogeneous soft tissue in subcutaneous plane. There was no evidence of arteriovenous fistula. Patient was already investigated with CT Brain earlier and the report suggested migrational abnormality with gliosis in the lateral wall of lateral ventricle. Chromosomal analysis showed normal 46XX karyotype. Blood investigations including hemogram, liver function
test, renal function tests and coagulation profile was normal.

**Discussion:**

Klippel-Trenaunay Syndrome was first described by two French doctors, Klippel and Trenaunay in 1900. It is a triad of vascular malformation, venous/lymphatic varicosity and soft tissue and bony hypertrophy. Haemangioma are often apparent at birth or by second week of age. Capillary haemangioma are the most common type and are called port wine stains due to its red and purple colour. If large enough, cutaneous haemangioma may cause sequestration of platelets, leading to Kasabach-Merritt syndrome, a type of consumptive coagulopathy. The haemangioma often overlies the vascular malformation. Varicose veins result from damaged or defective valves in a vein. Vein gets damaged when the smooth muscle in the wall of vein weakens and the valves cannot support the weight of blood. Bone and soft tissue hypertrophy is a result of increased growth. In many cases, limb length is affected. In most cases, the girth of the limb is larger, although atrophy is seen in some patients. The lower limb is involved in about 95% of patients while upper limb involvement is seen in 5%. Rarely only the trunk is involved. It affects males more than females.

When Klippel-Trenaunay Syndrome is associated with arteriovenous fistula, it is known as Klippel-Trenaunay-Weber Syndrome. A series of 252 patients with KTS was studies 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. Atypical veins, including lateral veins and persistent sciatic vein, occurred in 182 patients.

Other less common manifestations of KTS include thromboembolic episodes, thrombophlebitis, Kasabach-Merritt syndrome, haematuria, rectal or colonic bleeding, and vaginal, vulval or penile bleeding in children with visceral and pelvic haemangioma. Kasabach-Merritt syndrome can present as high output failure. Neoplastic risk is not increased in KTS. Although the cause of KTS is still unknown, it is hypothesized that it is caused by a mesodermal abnormality during foetal development leading to vascular and soft tissue malformations in the affected limb. Mc Grory & Amadio believed that an underlying mixed mesodermal and ectodermal dysplasia was responsible for development of KTWS.

Klippel-Trenaunay Syndrome might develop due to a single gene defect. Rarely it can be inherited as an autosomal dominant trait. Whelan et al. reported a case of a girl with KTW syndrome associated with a reciprocal translocation: t(5;11)(q13.3;p15.1). The de novo translocation t(8;14)(q22.3;q13) has also been reported. The association between the angiogenic factor gene AGGF1 and KTS appears to be significant. No definitive treatment is possible for KTS. Imaging studies like contrast enhanced MRI, Ultrasonography and Doppler study may be needed for diagnosis and to find out the extent of lesion that helps in planning the interventions if indicated. Treatment is indicated to reduce the symptoms and risk of complications. Active intervention needs to be attempted only for localized lesion or in presence of serious complications like bleeding or cardiac failure. Options available to treat the symptoms of KTS are surgery, sclerotherapy, and compression therapy. Laser treatment of the haemangioma can be effective in lightening the colour of the port-wine stain. Currently, the flash lamp-pumped pulsed dye laser is the treatment of choice in vascular lesions. It is also indicated in the
presence of ulceration. When treated with laser, ulcers heal more quickly. Laser treatment is most effective when performed early. Multiple sittings are required to achieve the desired effect.

Different surgical interventions for varicose veins include vein ligation, vein stripping, vein resection, and amputation. Vein ligation is a procedure which clamps or ties off a section of veins. It prevents blood flow through the damaged veins and promotes blood flow through normal veins. Vein stripping uses a metal wire to remove varicosities from within the damaged vein. Lindenauer SM suggested that the deep venous system is atretic in KTW syndrome, so stripping of varicose veins is unwise. Vein resection or excision removes a section of damaged veins from the body. Endovenous Thermal Ablation is a newer version of ligation and stripping of veins. In the procedure a laser or high frequency radio waves are given to produce intense heat locally in the varicose vein. It is less painful with fast recovery. In some cases, amputation of involved digits or extremity has to be done. Sclerotherapy can be done by using chemicals like sotradecol, ethanolamine, and absolute ethyl alcohol. It stops the blood flow through defective veins by causing inflammation in the inner lining of the veins. The vein later collapses and absorbed by the body. Debulking procedures have limited use and may damage venous and lymphatic structures leading to increased edema in the affected limb. Compression garments are indicated for chronic venous insufficiency, lymphedema, recurrent cellulitis and recurrent bleeding from capillary or venous malformations. Compression garments also protect the limb from trauma. Various compression garments available are compression socks, elastic wraps, neoprene wraps and other more complex devices. Many studies have given positive results in patients using compression therapy. Cellulitis and thrombophlebitis can be managed with analgesics, elevation, antibiotics, and corticosteroids. Radiotherapy may help to induce regression of hemangiomas though the results are slow to develop. Complications due to hemangioma include ulceration, bleeding, and secondary infection.

Complications of varicosities include paresthesia, ulcers, dermatitis, pulmonary embolism, thrombophlebitis, haemorrhage, and cellulitis. Hypertrophy of a limb may lead to vertebral scoliosis and gait abnormalities. It can cause degenerative joint disease also. Regarding limb hypertrophy, heel inserts are generally sufficient for limb length discrepancies of 1.5cm or less. If projected leg length discrepancy exceeds 2.0 cm at skeletal maturity, it can be treated by epiphysiodesis in the growing child. Patients with KTS should be monitored at least annually and more often if clinically indicated. Stable disease can be followed clinically. If the disease progresses, imaging studies should be performed and medical or surgical intervention should be pursued if indicated.
Figure 1 & 2: Showing hypertrophy of left leg, foot and two toes with left facial hemi hypertrophy

Figure 3 & 4: Showing port wine stains on left foot and lumbo-sacral region

Figure 5: Showing improper dentition

Figure 6: Showing normal radiograph of bilateral lower limb without any evidence of bony hypertrophy on the left

References:
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