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Case Report:

Klippel–Trénaunay syndrome –Benign, Cosmetic Disease

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Abstract:

Klippel-Trenaunay Syndrome (KTS) is a sporadic disorder characterized by the triad of vascular malformation (capillary hemangioma 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.

Introduction:

Klippel-Trénaunay syndrome (KTS or KT), formerly known as Klippel-Trénaunay-Weber syndrome and sometimes as angioosteohypertrophy syndrome, hemangiectatic hypertrophy, is a rare congenital medical condition in which blood vessels and/or lymph vessels fail to form properly. The three main features are nevus flammeus (Port-wine stain), venous and lymphatic malformations, and soft-tissue hypertrophy of the affected limb.

Klippel Trenaunay Syndrome is a rare mesodermal abnormality. It has three major features:

1) vascular naveus, 2) hypertrophy of soft tissue and bony overgrowth, and 3) varicose veins. It has a wide spectrum of presentation, from truncular involvement to extratruncular and from infiltrating to limited form of presentation. Klippel Trenaunay Syndrome must be differentiated from the Parkes-Weber syndrome. Both are characterized by overgrowth limbs. Both share similar clinical features, but represent separate clinical entities with different pathogenesis and natural history. While the key vascular components of Klippel Trenaunay Syndrome is capillary-lymphatic-venous malformation, the Parkes-Weber syndrome comprises a congenital persistence of multiple microscopic arteriovenous fistulas and varicosities. However, the management and prognosis of these two syndromes are distinctly different. The diagnosis of KTS is merely clinical and the treatment is conservative unless complications occur. We are presenting a case of classical KTS with elements of hypotrophy resembling myotonic dystrophy features with brief review of KTS literatures.

Case Report:

Ten years old male child presented in the OPD with history of progressive enlargement of right lower limb since birth (Fig. I). On examination, he had marked hypertrophy of the right lower limb. The measurements of both the thigh and legs were different, thigh were 31cms and 35.5 cms left and right and were 22 cms and 24.5 cms in the left and right legs respectively. There was spinal hypertrophy present in the lumbar region(Fig II). There was a large port wine stain on right gluteal area and posterior aspect of thigh (Fig. III). Doppler of the local area showed dilated great saphenous vein with grade 3

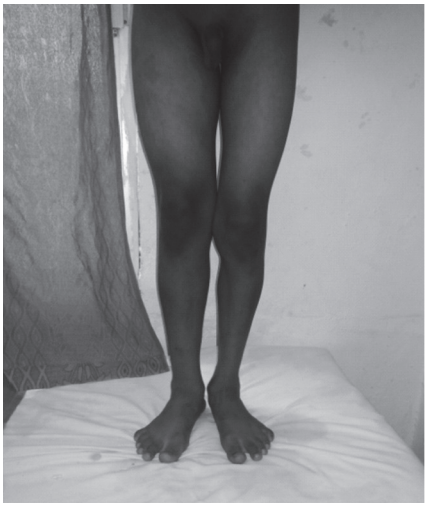

Fig. I

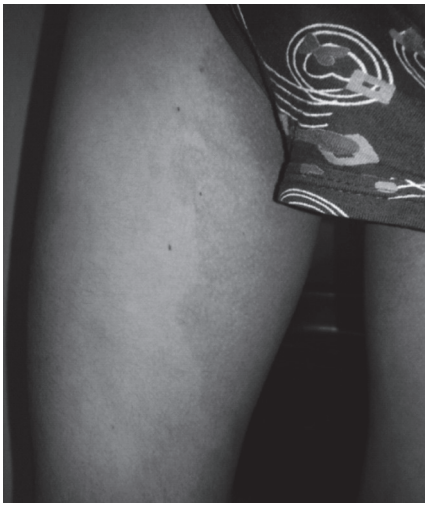

Fig. II

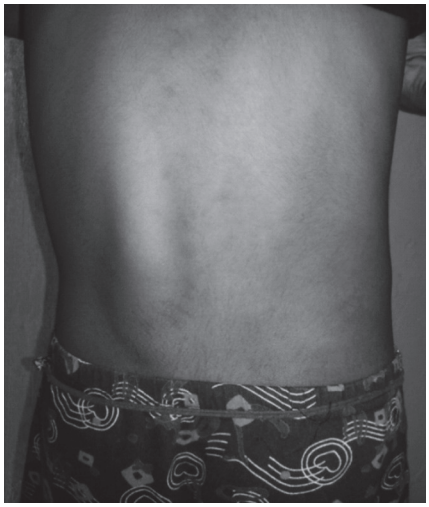

Fig. III

reflux and superficial varicosities in lower limbs with soft tissue edema in lower limb.These findings confirmed that it was a predominantlysoft tissue swelling containing pauci vascular hemangioma. As the child did not have any complication and the routine investigations were normal,so parents were reassured and appraised of the possible complications and were advised to come for regular follow up for limb length monitoring.

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 (Klippel & Trenaunay,1900). Hemangiomas are often apparent at birth or by second week of age (Samuel & Spitz, 1995). Capillary hemangiomas are the most common type and are called port wine stains due to its red and purple coluor. If large enough, cutaneous hemangiomas may cause sequestration of platelets, leading to Kasabach-Merritt syndrome, a type of consumptive coagulopathy. The hemangioma 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% (Phadke, 2009). 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 (KTWS; Weber,1907).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. Atypical veins, including lateral veins and persistent sciatic vein, occurred in 182 patients (72%; Jacob et al, 1998).Other less common manifestations of KTS include thromboembolic episodes, thrombophlebitis, Kasabach-Merritt syndrome, haematuria, rectal or colonic bleeding, vaginal, vulval or penile bleeding in children with visceral and pelvic haemangiomas. 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 fetal development leading to vascular and soft tissue malformations in the affected limb (Baskerville et al, 1985). McGrory & Amadio (1993) 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 (Happle,1993). Rarely it can be inherited as an autosomal dominant trait (Cebllos-Quintal et al, 1996). Whelan et al (1995) 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 (Wang et al,2001). The association between the angiogenic factor gene AGGF1 and KTS appears to be significant (Hu et al, 2008).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 intended 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 hemangioma can be effective in lightening the color of the port-wine stain. Currently, the flashlamp-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 (1965) 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 have 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 (Stringel & Dastous, 1987).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, hemorrhage, 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 1.5 cm 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.

Conclusion:

This is usually a benign condition were the problem is only cosmetic in nature. The complication is not common. Only symptomatic treatment is given. The patient is counselled and reassured. Regular follow up is necessary.

Bibliography:

1. Zea MI, Hanif M, Habib M, Ansari A (2009) Klippel-Trenaunay Syndrome: a case report with brief review of literature. J Dermatol Case Rep 3: 56-59.
2. Fait G, Daniel Y, Kupferminc MJ, Gull I, Peyser MR, et al. (1996) Klippel-Trénaunay-Weber syndrome associated with fetal growth restriction. Hum Reprod 11: 2544-2545.
3. Gloviczki P, Driscoll DJ (2007) Klippel-Trenaunay syndrome: current management. Phlebology 22: 291-298.
4. Samuel M, Spitz L: Klippel-Trenaunay syndrome: clinical features, complications and management in children.British Journal of Surgery, 1995; 82(6):757-761.
5. Weber FP, A.f.i.c.w.h.o.l.a.h. BJD (1907) Angioma formation in connection with hypertrophy of limbs and hemihypertropy. Br J Dermatol 19: 231-235.
6. Mazoyer E, Enjolras O, Laurian C, Houdart E, Drouet L (2002) Coagulation abnormalities associated with extensive venous malformations of the limbs: differentiation from Kasabach-Merritt syndrome. Clin Lab Haematol 24: 243-251.
7. Alomari AI (2009) A truly unusual overgrowth syndrome: an alternative diagnosis to Klippel-Trénaunay-Weber syndrome. Intern Med 48: 493-494.
8. Oduber CE, Khemlani K, Sillevis Smitt JH, Hennekam RC, van der Horst CM (2010) Baseline Quality of Life in patients with Klippel-Trenaunay syndrome. J Plast Reconstr Aesthet Surg 63: 603-609.
9. Phadke SR: Klippel Trenaunay syndrome. Atlas of Genetics and Cytogenetics in Oncology and Haematology, 2009;13 (2):153-155.
10. Danarti R, König A, Bittar M, Happle R (2007) Inverse Klippel-Trenaunay syndrome : review of cases showing deficient growth. Dermatology 214: 130-132.
11. Suchitra G, Madhu.R, Srinivasan MS: Klippel Trenaunay Syndrome. e-Journal of the Indian Society ofTeledermatology. 2008; 2(4):7-14.
12. Whelan AJ, Watson MS, Porter FD, Steiner RD: Klippel-Trenaunay-Weber syndrome associated with a 5:11 balanced translocation. American Journal of MedicineGenetic, 1995;59(4):492-494
13. Wang Q, Timur AA, Szafranski P, Sadgephour A, Jurecic V, Cowell J, Baldini A, Driscoll DJ: Identification and molecular characterization of de novo translocation t(8;14)(q22.3;q13) associated with a vascular and tissue overgrowth syndrome. Cytogenetic & Cell Genetic,2001; 95(3-4): 183-188.
14. Stringel G, Dastous J: Klippel-Trenaunay Syndrome and other cases of lower limb hypertrophy: Pediatric surgical implications. Journal of Pediatric Surgery, 1987;22(7) :645-650.