Case Report

Klippel -Trenaunay Syndrome: A Case Report

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Abstract

We report a case of a 15 years old lady with Klippel- Trenaunay syndrome who came with soft tissue swelling of the left limb since birth. The lady was initially mis-diagnosed and treated as lymphatic filariasis by a local health practitioner. The case is reported because of its rare presentation. A brief review of literature is also done.

Keywords: Klippel -Trenaunay Syndrome, capillary- venous malformation.

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Introduction

Klippel-Trenaunay syndrome(KTS) is a cutaneous capillary malformation on a limb in association with soft tissue swelling, with or without bony hypertrophy. It is classified as slow- flow complex combined capillary- venous malformation (CVM) or capillary- venous- lymphatic malformation (CVLM). Lesions are present at birth and in approximately 75% of patients they appear before 10 years of age.[1]

It is a sporadic condition with multifactorial inheriitance.[2] Its cause continues to be investigated, although theories abound. There is no predilection for gender or any particular ethnicity, and it appears more frequently at birth, childhood or adolescence. We report a case of 15-year-old lady with this syndrome.

Case history

A 15 years old female presented with the complaints of swelling of left lower limb since birth. The swelling had increased in size gradually. The parents also had noticed multiple non itchy red patches over the same limb since birth. Swelling of limb was always associated with episodic fever and pain. There was history of similar non itchy red patches on trunk of her mother and sister. On examination, there was gross hypertrophy of the left leg with 15x15 cm and 10x8 cm veruccous plaques on the left shin and dorsum of left foot respectively (Figure 1). There was fissuring and haemorraghic crust over the plaque. In addition, there were multiple port wine stains of varying sizes extending from lateral aspect of the left iliac crest region to the lateral side of left knee joint(Figure 2). Multiple angiokeratomas were seen over the surface of these port wine stains. The right lower limb was normal. There was no ulceration, varicosity, orofacial abnormality, dental abnormality, spina bifida, polydactyly, oligodactyly, syndactyly or visible pulsatile mass.

The X-ray of the left lower limb revealed subcutaneous swelling, loss of subtarsal joint space with some deformity of the calcaneal subtarsal articulation.
The peripheral arterial and venous doppler study showed marked thickening of the skin and subcutaneous tissue in the leg and foot and the vessels could not be traced in the distal part of the leg. Other hepatic, renal, and hematological investigations were within normal limits.

Discussion

In the case reported, the patient showed port-wine stains in the lower limb. Varicose veins were not visible on physical and imaging studies of lower limbs. A slight elongation of the patient's left hemibody in length and gross increase in circumference of left lower limb was visible. Hypertrophy of bone was not seen in this case. Doppler study showed no arteriovenous malformation. Based on the clinical findings of port wine stain, a capillary malformation and soft tissue hypertrophy of lower limb since birth without AV malformation, a diagnosis of Klippel Trenaunay syndrome was made. The verrucous plaque observed in the limb could be explained due to repeated episode of lymphangitis in a grossly hypertrophic limb of long duration. Klippel-Trenaunay Syndrome was first noted in a 1900, by French physicians Klippel and Trenaunay.[3] The cause of this syndrome is unknown. There are two theories that have been argued by the medical community. The first argument is that Klippel-Trenaunay Syndrome is a mesodermal abnormality during fetal development.[4] The mesodermal abnormalities cause vascular and soft tissue malformations in the affected limb. The other argument states that Klippel-Trenaunay Syndrome is caused by gene mutation. Although heredity does not appear to be the cause of Klippel-Trenaunay Syndrome, it is a possible option .[5]

Although KTS is a sporadic condition, studies report familial cases of KTS that have not been inherited from a Mendelian pattern, thus suggesting a multifactorial inheritance.[2] Later studies conducted by Happle suggest that the inheritance of a single abnormal gene could explain the development of this syndrome, as well as the occurrence of sporadic and familial

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cases. [6]

The full-blown classical syndrome of KTS has a triad of congenital mesodermal abnormalities characterized by port-wine stain, venous varicosities, and hypertrophy of soft tissue and/or overgrowth of bone of one or more limbs.[7] However, all patients do not manifest the full triad. In a study by Gloviczki et al[3], port-wine stain was present in 95% of cases, hypertrophy of the soft tissues or bones in 93%, and varicosities in 76% of 144 cases of KTS. Moreover, unusual variants of the syndrome are often described, such as limb hypotrophy.[8,9]

Most patients show the three symptoms of the clinical syndrome, and hemangioma is often the first to appear. Port-wine stain or flat hemangioma is a vascular malformation present at birth and that does not show tendency toward involution. It is often unilateral and segmented, never crossing the midline. It increases in proportion to the child's growth and may involve any part of the body, although face and cervical region are the most commonly affected areas. Lesions may be light pink in infancy and become progressively darker (dark red) as the child ages.[10] Portwine stain may be limited or extend to deeper areas of skin, including bones, muscles, and organs, worsening the prognosis of the disease.

Varicose veins observed in patients with the syndrome may be noticed in early infancy, but they generally become prominent in a later stage and progress until adolescence.[11] They are a large and lateral vein, which starts on the foot or leg, proximally, and extends until the buttocks or gluteus region. These areas may remain stable or enlarge gradually, causing pain, lymphedema, thrombophlebitis, and ulcers. Hypertrophy is the third symptom to appear in the syndrome and it can be secondary to length increase (bone involvement) and/or circumference increase (soft tissue involvement). It can be observed at birth and progresses during the first years of life. In adolescence, when the child's growth cycle period has finished, the limb will stop growing. 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 non syndromic capillary malformations of the skin.[11]

There is no cure for this disorder. Therapeutic objectives seek to improve the patient's condition and treat the consequences of severe lesions and length discrepancy. Treatment of port-wine stains is done with *pulsed dye laser* therapy. It is best to start treatment early because younger children require fewer sessions and show more favorable results. Treatment 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.[12]

When varicose veins are present, compression stockings are recommended for venous insufficiency. Surgical treatment is only recommended in symptomatic cases of superficial varicose veins.[13]

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.

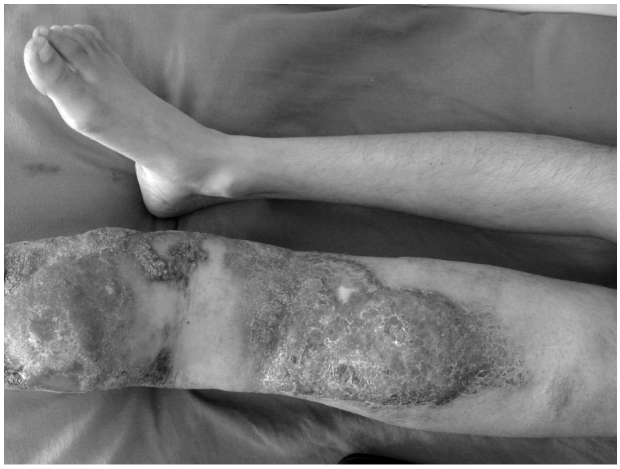

Figure 1 : Soft tissue swelling of left leg

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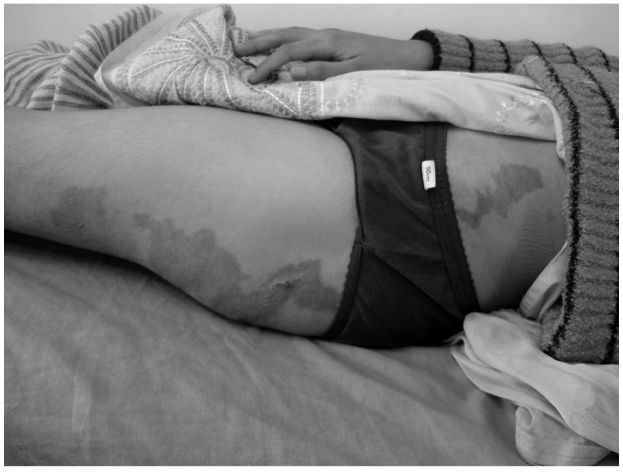

Figure 2 : Port wine stain extending from left iliac region to the Postero-lateral aspect of left thigh

Conclusion

In conclusion, KTS is a rare syndrome. Patients diagnosed with KTS must be evaluated once a year or more frequently, based on clinical recommendation, in order to keep the disease under control. If the disorder progresses, imaging studies should be done and surgical intervention should be considered for the correction of lower limb length discrepancy. Research in genetics should be encouraged so that in the future we may be able to understand the etiology of this disease.

References

1. Favorito LA. Vesical Hemangioma in patient with Klippel-Trenaunay-Weber syndrome. *J Urol*. 2003;29:149-50.

2. Aelvoet GE, Jorens PG, Roelen LM. Genetic aspects of the Klippel-Trenaunay syndrome. *Br J Dermatol*. 1992;126:603-7.

3. Gloviczki, P., L.H. Hollier, R.L. Telander, B. Kaufman, A.J. Bianco, and G.B. Stickler. Surgical Implications of Klippel-Trenaunay Syndrome. *Annals of Surgery.* 1983 ;197(3):353-360

4. Baskerville, P.A. J.S. Ackroyd, and N.L. Browse. The Etiology of the Klippel-Trenaunay syndrome. *Annals of Surgery.*1985; 202(5):624-7.

5. Nielson, J.R. and E.H. Tschen. Klippel-Trenaunay-Weber Syndrome. *Cutis.* 1987; 40(1):51-53.

6. Happle R. Klippel-Trenaunay syndrome: is it a paradominant trait? *Br J Dermatol*. 1993; 128:465-6.

7. Atiyeh BS, Musharrafieh RS. Klippel–Trenaunay-type syndrome: aneponym for various expressions of the same entity. *J Med* 1995;26:23.

8. Irlbacher K, Hoffmann KT, Behse F et al. Variant of Klippel–Trenaunay syndrome. A case report. *Rontgenpraxis* 2000; 52:312–16.

9. Moss C, Robson BJ. Haemangiectatic hypotrophy associated with Perthe's disease. *Clin Exp Dermatol* 1988; 13:237– 60.

10. Gontijo B, Pereira LB, Silva CMR. Malformações Vasculares. *An Bras Dermatol*. 2004; 79:7-25.

11. Garzon MC, Huang JT, Enjolras O, Frieden IJ. Vascular Malformations/Part II: Associated syndromes. *J Am Acad Dermatol*. 2007;56:541-64.

12. Richards KA, Garden JM. The pulsed dye laser for cutaneous vascular and nonvascular lesions. *Sem Cutan Med Surg*. 2000;19:276-86.

13. Gloviczki P, Stanson AW, Stickler GB, Johnson CM, Toomey BJ, Meland NB, et al. Klippel Trenaunay syndrome: the risks and benefits of vascular interventions. *Surgery.* 1991;110:469-79.