A rare presentation of Klippel-Trenaunay syndrome

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ABSTRACT

The Klippel-Trenaunay syndrome (KTS) is a congenital disorder characterized by capillary malformation, varicosities and bony and soft tissue hypertrophy. This disease has several morbidities like bleeding, deep venous thrombosis, embolic complications and in some cases enlargement of limb that may require amputation. Vascular malformations are segmented and never cross midline. However, we came across a case, a 45-year-old male, who presented with varicosity of veins and deformity of left lower limb besides cavernous hemangiomas (port-wine stains) scattered all over his face, chest, back, gluteal region, groin and legs since birth. Multiple paravertebral soft tissue masses and bladder hypertrophy were also noted due to involving neurofibromatosis. Simultaneous occurrence of KTS and neurofibromatosis is rarely seen in clinical practice.

Key words: Klippel-Trenaunay syndrome, limb, paravertebral soft tissue mass hypotrophy

INTRODUCTION

Klippel-Trenaunay syndrome (KTS) is characterized by triad of port-wine stain, varicose veins, and bony and soft tissue hypertrophy. Herein we report a case of KTS with a rare presentation.

CASE REPORT

A 45-year-old male presented with an ulcer over the left ankle. On examination, dilated tortuous veins were present all over the left lower limb including gluteal region. There was associated muscular atrophy of left leg and deformity of left great toe since birth. Cavernous hemangiomas were present on his face, chest, back, gluteal region, groin and legs and were observed mostly on the left side [Figures 1-3]. Local temperature over the affected surface was not raised. Subcutaneous inguinal swellings were present that were non-tender, soft and irreducible. They were neither compressible and nor pulsatile. There was no history of hematemesis, melena, hematochezia or hematuria. He had no family history of similar disorder and had been asymptomatic till he developed ulcer over left ankle 4 months ago. After initial physical evaluation, investigations were advised. A routine hematology revealed normal results. Doppler study revealed incompetence of venous valves at all level in the left lower limb. There was no evidence of arterio-venous fistula or deep venous thrombosis. However, the arteries showed atherosclerotic changes, with sub-cutaneous edema all over the left lower limb. Abdominal and pelvic ultrasound revealed subcutaneous soft tissue swellings in the inguinal and gluteal region and abnormally enlarged urinary bladder. Computerized tomographic scan of the chest and abdomen revealed paravertebral soft-tissue attenuated masses from D_1 to D_{10} [Figure 4].

DISCUSSION

About a century ago, two French physicians Maurice Klippel and Paul Trenaunay described two patients with hemangiomatous lesions of the skin associated with asymmetric soft tissue and bone hypertrophy, and coined the term “naevus variqueux osteohypertrophique.”[1] Years later, Frederick Parks Weber noted the occurrence of these findings in association with arteriovenous fistulas. The related Klippel-Trenaunay-Weber syndrome describes those individuals who have an arteriovenous malformation as a component of their syndrome. Various case reports of KTS are present in world literatures. Still, incidence and genetic predisposition of this rare disease has not been established. The classic clinical triad

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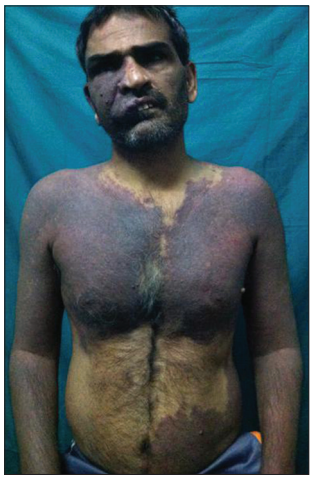

Figure 1: Port wine stain involving face, chest, both upper limbs

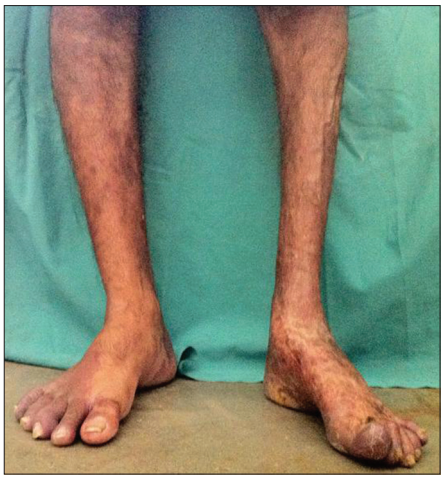

Figure 2: Atrophy of muscles of left leg with deformity of great toe

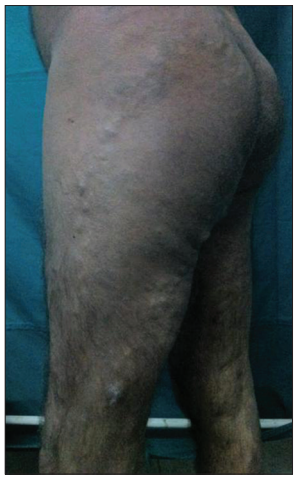

Figure 3: Varicosity of veins over buttock and lateral aspect of thigh

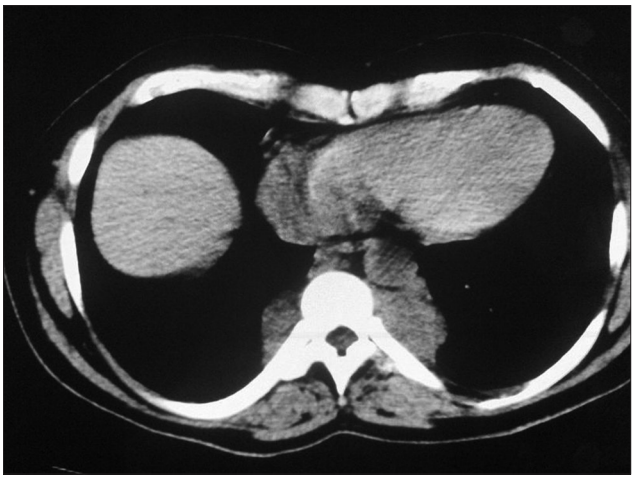

Figure 4: Computer tomography lower chest showing paravertebral soft tissue mass

includes capillary malformations (port wine stain), a usually longer and larger extremity because of soft tissue and bone hypertrophy, and atypical mostly lateral superficial varicosity. Patients with at least two of the three cardinal features have been classified as having an incomplete form of KTS.

KTS affects males more than females and overall incidence is 2-5/100,000. It generally affects only one extremity. Lesions are usually present since birth and in approximately 75% of patients they appear before 10 years of age.[2] The origin of this syndrome continues to be investigated, and many theories were discussed. 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 cases.[3,4] Although KTS is a sporadic condition, studies report familial cases of KTS that have not been inherited in a Mendelian pattern, suggesting a multifactorial inheritance.

KTS was initially described in 1900 as a triad of cutaneous capillary hemangiomas, bony and soft-tissue hypertrophy, and varicosity of veins.[5] The disorder usually affects one body segment and has several other clinical manifestations. Most patients often show these three components 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. Hemangiomas 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 at a later stage and progress until adolescence. These are large lateral veins that start on the foot or leg proximally and extend upto the gluteal region. These areas may remain

stable or enlarge gradually, causing pain, lymphedema, thrombophlebitis, and ulcers. Vascular malformations are postulated to be the cause of hypertrophy.

Hypertrophy is the third symptom to appear in the syndrome and it can be secondary to increase in length of bones and/or increase in circumference due to soft-tissue involvement. It can be observed at birth and progresses during the first year of life. In adolescence, when the child's growth cycle is complete, the limb will stop growing.

Vascular malformations involving the gastrointestinal and genitourinary tracts have been reported and can be a significant source of morbidity and even mortality. Patients with vascular malformations of the bladder frequently have associated rectosigmoid or other pelvic organ involvement. Rectal and bladder hemorrhage are serious complications of pelvic vascular malformations and have been reported in 1% of cases. Involvement of the gastrointestinal tract may be more common in KTS than previously believed (occurring in perhaps as many as 20% of patients) and may go unrecognized in patients without overt symptoms.[7-9]

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.[6] 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.

The lower limb is the site of malformation in approximately 95% of patients. When found on the trunk, the malformation rarely crosses the midline. The hypertrophy involves the length as well as the circumference of the involved extremity and is caused by local hyperemia and venous stasis secondary to the vascular anomaly.[10] However, in the present case we observed that capillary malformations were extensively present involving both halves of the body, with obvious hypotrophy of left lower limb. The exact cause of limb atrophy is unknown. However, in this it may be attributed to regional fatty atrophy and atherosclerosis of vessels. Soft-tissue mass was present subcutaneously and in paravertebral area and urinary bladder was abnormally large and highly placed, probably due to neurofibromatosis. Simultaneous occurrence of KTS and neurofibromatosis is rarely seen in clinical practice.

REFERENCES

1. Klippel M, Trenaunay P. Du naevus variquex osteohypertrophique. Arch Gen Med 1900;3:641-72.
2. Garzon MC, Huang JT, Enjolras O, Frieden IJ. Vascular malformations: Part I. J Am Acad Dermatol 2007;56:353-70.
3. Garzon MC, Huang JT, Enjolras O, Frieden IJ. Vascular malformations. Part II: Associated syndromes. J Am Acad Dermatol 2007;56:541-64.
4. Tian XL, Kadaba R, You SA, Liu M, Timur AA, Yang L, *et al*. Identification of an angiogenic factor that when mutated causes susceptibility to Klippel-Trenaunay syndrome. Nature 2004;427:640-5.
5. Berry SA, Peterson C, Mize W, Bloom K, Zachary C, Blasco P, *et al*. Klippel-Trenaunay syndrome. Am J Med Genet 1998;79:319-26.
6. Jacob AG, Driscoll DJ, Shaughnessy WJ, Stanson AW, Clay RP, Gloviczki P. Klippel-Trénaunay syndrome: Spectrum and management. Mayo Clin Proc 1998;73:28-36.
7. Servelle M, Bastin R, Loygue J, Montagnani A, Bacour F, Soulie J, *et al*. Hematuria and rectal bleeding in the child with Klippel and Trenaunay syndrome. Ann Surg 1976;183:418-28.
8. Wilson CL, Song LM, Chua H, Ferrara M, Devine RM, Dozois RR, *et al*. Bleeding from cavernous angiomatosis of the rectum in Klippel-Trenaunay syndrome: Report of three cases and literature review. Am J Gastroenterol 2001;96:2783-8.
9. Schmitt B, Posselt HG, Waag KL, Müller H, Bender SW. Severe hemorrhage from intestinal hemangiomatosis in Klippel-Trenaunay syndrome: Pitfalls in diagnosis and management. J Pediatr Gastroenterol Nutr 1986;5:155-8.
10. Hale EK. Klippel-Trenaunay syndrome. Dermatol Online J 2002;8:13.

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