

Klippel- Trenaunay syndrome with multiple limb involvement – A case report

Konsam Biona Devi¹, Dhanaraj Singh Chongtham², Linda Marangmei³, Kamble Sadanand^{4*}, Anand Sonwane⁵

^{1,4,5}PG Student, ²Professor and Cardiologist, ³Senior Resident, Department of Medicine, Regional Institute of Medical Sciences Imphal, Manipur, INDIA.

Email: sadakamble@gmail.com

Abstract

Klippel-Trenaunay syndrome is a rare syndrome characterised by a triad of port-wine stain, varicose veins and bony and/or soft tissue hypertrophy involving extremity. Klippel-Trenaunay syndrome generally affects a single extremity. A case of Klippel-Trenaunay syndrome with multiple limb involvement and macrodactyly is reported.

Keywords: Klippel-Trenaunay syndrome, naevus vasculosus osteohypertrophicus, port-wine stain, bony hypertrophy, soft tissue hypertrophy, venous varicosities.

*Address for Correspondence:

Dr. Kamble Sadanand, PG Student, Department of Medicine, Regional Institute of Medical Sciences Imphal, Manipur, INDIA.

Email: sadakamble@gmail.com

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INTRODUCTION

Klippel-Trenaunay syndrome is a rare syndrome characterised by a triad of port-wine stain, varicose veins and bony and/or soft tissue hypertrophy. Diagnosis is purely on clinical examination. It can present in wide varieties ranging from a simple asymptomatic disease to a life threatening embolism.

CASE HISTORY

A 32 year old female patient presented to medicine out-patient department with complaints of dysphagia,

dysphonia and bloodish discharge from her left ear. General examination was revealing port wine stain over lower half of right arm and extensor aspect of right forearm (Figure 1). Limb discrepancy was present, in upper limb right>left (limb length discrepancy 3cm), in lower limb left>right (limb length discrepancy 2cm). Soft tissue hypertrophy was also present in right upper limb, right infra mammary, right infra scapular region and left lower limb. Macrodactyly was present in second toe of both the foot. Superficial veins were seen over both the lower limbs. Left ear was showing bleeding on otoscopic examination. Pure tone audiometry was showing minimal sensory neural hearing loss. Routine haematological investigations were normal. Upper G I endoscopy was done for evaluation of dysphagia, but it was normal. CT pulmonary angiography was not showing any A-V malformations. In venous colour Doppler of right upper limb and left lower limb, there was no clot or incompetence of the perforators. Patient was treated conservatively, advised for regular follow up annually and to monitor for any complication of haemangiomas, varicosities and hypertrophy of limb.



Figure 1



Figure 2



Figure 3



Figure 4

DISCUSSION

In 1900 Klippel and Trenaunay first described this syndrome in two patients presenting with a port-wine stain and varicosities of an extremity associated with hypertrophy of the affected limb's bony and soft tissue. They termed the syndrome "naevus vasculosus osteohypertrophicus." The exact etiology of this syndrome is not known. McGrory and Amadio believe that an underlying mixed mesodermal and ectodermal dysplasia is likely responsible for the development of Klippel-trenaunay weber syndrome¹. Most cases are sporadic, but few cases in literature reports an autosomal dominant inheritance². It affects females and males equally. In Klippel-Trenaunay syndrome capillary haemangioma or port-wine stain are most common type of malformation. Haemangiomas is often noted on the lateral aspect of the limb. They possess neither a proliferative nor a regressing phase. Hemangioma depth is variable. It may be limited to the skin or extend deeper to subcutaneous tissue, including muscle and bone. Visceral organs, such as the pleura, the spleen, the liver, the bladder, and the colon may also be affected. Visceral organ involvement portends greater morbidity secondary to internal hemorrhage that may manifest as hematuria or hematochezia. The venous malformations frequently present as persistence of embryonic veins. The Klippel-Trenaunay vein is a large, lateral, superficial vein seen in 68-80% of patients³. The vein is usually thick walled and strong, it is located immediately under the skin and it is incompetent along its entire length due to the absence of venous valves. Drainage is either into a lateral branch of the profunda femoris vein or into the internal iliac vein⁴. Another embryonic vein sciatic vein has also been described in Klippel-Trenaunay syndrome⁵. Varicose veins and venous malformations can involve abdominal and pelvic organs. Genitourinary manifestations may present as intrapelvic and retroperitoneal vascular malformations. Bleeding from capillary or venous malformations or persistent embryonic veins may occur through defects in the skin or mucosa, or the patient can have intramuscular or retroperitoneal haematoma, haematuria, rectal bleeding, intracerebral or intraspinal

haemorrhage. GI tract involvement may be more common in Klippel-Trenaunay syndrome than previously believed because most of cases remain unrecognized without overt symptoms⁶. Limb hypertrophy may occur at birth or within first years of life. It can be secondary to bony hypertrophy (increase in length) or soft tissue hypertrophy (increase in girth). Rarely, the involved limb may be atrophied rather than hypertrophied⁷. Limb discrepancy can cause gait disturbances. Other features include lymphatic obstruction, spina bifida, hypospadias, polydactyly⁸, syndactyly, oligodactyly, macrodactyly, hyperhidrosis, hypertrichosis, paresthesia, decalcification of involved bones, dislocation of hip⁹. Gastrointestinal haemorrhage¹⁰, chronic venous insufficiency, stasis dermatitis, poor wound healing, ulceration, thrombosis, angiosarcoma, and emboli.^{11,12}

Diagnosis

Diagnosis is mainly clinical with thorough history and physical examination. Investigations are useful when complications are associated with it. Detailed colour duplex scanning, contrast venography of the venous system of the leg to establish patency, incompetence, thrombosis, arteriovenous shunting and any anomalies such as hypoplasia. Plain X-rays of the long bones (scanograms), CT scans are most helpful to measure bone length. MRI is helpful in differentiating bone, fat, muscle hypertrophy and lymphedema.

Management

Prevention of venous thromboembolism with anticoagulation or inferior vena cava filter is as important as prevention of repeated episodes of cellulitis and lymphangitis in those with associated lymphoedema or as treatment of the symptomatic vascular anomalies. In Klippel-Trenaunay syndrome, 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. Laser treatment is also indicated in the case of ulceration. Ulceration of hemangiomas can be painful and can impair functional abilities. When treated with laser, ulcers often heal more quickly. Laser treatment is most effective when performed early, as it can improve the long-term appearance of the port-wine stain and thereby

also improve function. Typically, many treatments are required to achieve the desired effect. Laser treatment only helps with the superficial component of the hemangioma.¹³ The absolute indications of treatment are haemorrhage, infections, acute thromboembolism or refractory ulcers. The management of Klippel-Trenaunay syndrome has been largely conservative. Compression therapy has been the mainstay of conservative treatment in the form of an elastic garment or compression bandage. This has been beneficial in managing both lymphedema and chronic venous insufficiency. Local wound care, compression dressings, special orthopaedic footwear and lifestyle modification may also be required to manage activities of daily living and improve the function of the limb. Debulking procedures have limited use and may damage venous and lymphatic structures, leading to increased edema in the affected limb. The potential risks and benefits must be carefully weighed before attempting surgical intervention. Radiotherapy has been reported to be of help in some cases of Klippel-Trenaunay syndrome. The radiation may help to induce regression of hemangiomas; however, the results can be slow to develop.¹⁴ Endovenous laser therapy of the greater saphenous vein is gaining support for the management of varicosities in the general public and in patients with Klippel-Trenaunay syndrome.¹⁵ This therapy has been used alone and in combination with other surgical interventions. It is a novel and minimally invasive approach for the management of some varicosities.

CONCLUSION

Klippel Trenaunay Syndrome is a rare presentation and multiple limb involvement is even rarer. Exact incidence and pathogenesis is still unknown. Diagnosis is purely clinical. Management is conservative with lifelong follow up. Appropriate multidisciplinary approach of management is required as disease affects multiple organs.

REFERENCES

1. McGrory BJ, Amadio PC. Klippel-Trenaunay syndrome: orthopaedic considerations. *Orthop Rev.* Jan 1993; 22(1):41-50. [\[PubMed\]](#)
2. Ceballos-Quintal JM, Pinto-Escalante D, Castillo-Zapata I. A new case of Klippel-Trenaunay-Weber (KTW)

3. Kim YW, Lee BB, Cho JH, et al. Haemodynamic and clinical assessment of lateral marginal vein excision in patients with a predominantly venous malformation of the lower extremity. *Eur J Vasc Endovasc Surg.* 2007; 33: 122-127. [\[PubMed\]](#)
4. Sernelle M. Klippel-Trenaunay syndrome: 768 operated cases. *Ann Surg.* 1985; 201: 365-373. [\[PubMed\]](#)
5. Cherry KJ, Gloviczki P, Stanson AW. Persistent sciatic vein: diagnosis and treatment of a rare condition. *J Vasc Surg.* 1996; 23: 490-497. [\[PubMed\]](#)
6. Cha SH, Romeo MA, Neutze JA. Visceral manifestations of Klippel-Trenaunay syndrome. *Radiographics.* 2005; 25: 1694-1697. [\[PubMed\]](#)
7. Paller AS, Manini AJ. Vascular disorders of Infancy and Childhood. Hurwitz. *Clinical Pediatric Dermatology.* 3rd edn, Elsevier Saunders, 2006; 307-344.
8. Sunar H; Halici U; Duran E. Klippel-Trenaunay syndrome associated with polydactyly. [\[PubMed\]](#)
9. Skourtis G, Lazoura O, Panoussis P, Livieratos L. Klippel-Trenaunay syndrome: an unusual cause of pulmonary embolism. *Int Angiol.* 2006; 25(3):322-6. [\[PubMed\]](#)
10. Pereda Marín RM; García Collada JC; Garrote Martínez AI; Miralles Serrano EM; Morales Aguilar JL. Anesthetic management of Klippel-Trénaunay syndrome and attendant gastrointestinal hemorrhage. A case report. *Minerva Anesthesiol.* 2007; 73(3):187-90. [\[PubMed\]](#)
11. Furness PD 3rd, Barqawi AZ, Bisignani G, Decter RM. Klippel-Trénaunay syndrome: 2 case reports and a review of genitourinary manifestations. *J Urol.* Oct 2001; 166(4):1418-20. [\[PubMed\]](#)
12. Ploegmakers MJ, Pruszczynski M, De Rooy J, Kusters B, Veth RP. Angiosarcoma with malignant peripheral nerve sheath tumour developing in a patient with klippel-trénaunay-weber syndrome. *Sarcoma.* 2005;9(3-4):137-40. [\[PubMed\]](#)
13. Spicer MS, Goldberg DJ, Janniger CK. Lasers in pediatric dermatology. *Cutis.* May 1995; 55(5):270-2, 278-80. [\[Medscape\]](#)
14. Yildiz F, Yilmaz M, Cengiz M, et al. Radiotherapy in the management of Klippel-Trénaunay-Weber syndrome: report of two cases. *Ann Vasc Surg.* Jul 2005;19(4):566-571. [\[WebLink\]](#)
15. Huang Y, Jiang M, Li W, Lu X, Huang X, Lu M. Endovenous laser treatment combined with a surgical strategy for treatment of venous insufficiency in lower extremity: a report of 208 cases. *J Vasc Surg.* Sep 2005; 42(3):494-501; discussion 501. [\[PubMed\]](#)

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