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Case Report

Klippel-Trenaunay Weber Syndrome-Report of 2 Cases

Rajput Chetan[1], Chaudhary Hemant[2], Shameela Aafreen[3], Gore Sanjay[4]
[1, 4]Assistant Professor, Department of Skin and VD, S.B.H Government Medical College Dhule-424001, Maharastra, India
[2]Associate Professor, Department of Surgery, S.B.H Government Medical College Dhule-424001, Maharastra, India
[3]Senior Resident, Department of Skin and VD, S.B.H Government Medical College Dhule-424001, Maharastra, India

***Corresponding author**
Rajput Chetan
Email: drchetanrajput@yahoo.com

Abstract: Klippel-Trenaunay Syndrome (congenital dysplastic angiopathy) is a congenital vascular disorder of unknown etiology.It is a sporadic disorder characterized by the triad of vascular malformation in the form of capillary hemangioma or port wine stain, venous varicosity of the lower limb and soft tissue and/ or bony hypertrophy. We report 2 case of Klippel-Trenaunay syndrome with review of literature.
Keywords: Klippel-Trenaunay syndrome, Capillary heamangioma, Port wine stain, Venous verocosities, Hypertrophy

INTRODUCTION

Klippel-Trenaunay syndrome (KTS) is a rare disorder with an incidence of 3-5/1,00,000 [1]. It is characterized by a triad of vascular malformations in the form of capillary heamangioma or port-wine stain, varicose veins, and bony and soft tissue hypertrophy. It most commonly affects only one limb at a time but multiple site involvement can also occur [2]. It's a congenital dysplastic disorder and approximately 75% of patients presented before 10 years of age [3]. The difference between KTS and Klippel-Trenaunay-Weber syndrome (KTWS) is that the latter includes significant arteriovenous malformations in the affected extremity [4]. Other names for Klippel-Trenaunay Syndrome are Angio-osteohypertrophy, Nevus varicosus osteohypertrophicus syndrome, Hemangiectasia hypertrophicus and Nevus verucosus hypertrophicans.

CASE REPORT

Case 1:

A 11 years female born of non-consangionous marriage presented with right lower limb hypertrophy (Fig. 1) and port-wine stains over right buttock and knee (Fig. 2) since birth. Histopatholgy of skin biopsy shows papillary dermis with dilated lymphatic channels surrounded by lymphatic infiltrates and stroma infiltrating lymphocytic cells, all suggestive of lymphangioma. Other routine investigations along with X-ray and Doppler studies revealed soft tissues and bony hypertrophy.

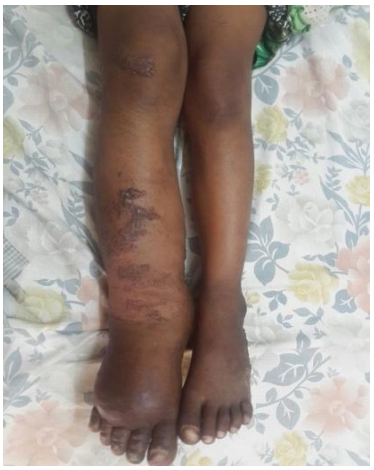

Fig. 1: Hypertrophic right lower limb with venous and lymphatic varicosities

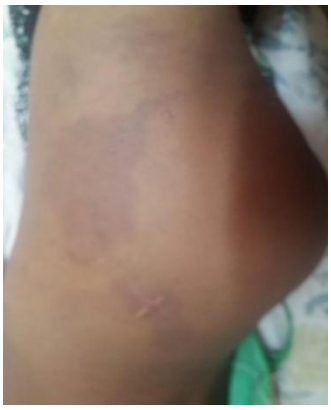

Fig. 2: Port wine stain over right buttock and knee

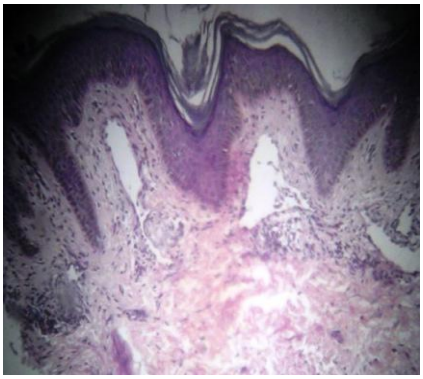

Fig. 3: Histopathology showing papillary dermis with dilated lymphatic channels surrounded by lymphatic infiltrates and stroma infiltrating lymphocytic cells, all suggestive of lymphangioma

Case 2

A 10 years old male child BONCM presented with hypertrophy of left lower limb (Fig. 4), dilated veins over postero-lateral aspects of left thigh and leg and port wine stains over left thigh, knee and leg (Fig. 5) since birth. Colour Doppler study of left lower limb showed varicose veins in postero-lateral aspects of entire thigh & leg. Radiological examination showed soft tissue swelling.

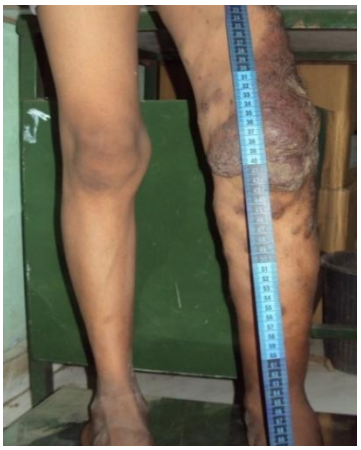

Fig. 4: Hypertrophic left lower limb with venous varicosities

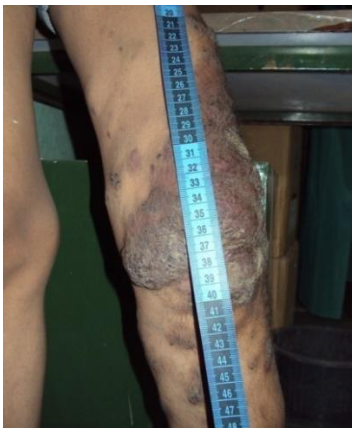

Fig. 5: Showing port wine stain with venous varicosity

DISCUSSION

Klippel-Trenaunay Syndrome was first described by two French doctors, Klippel and Trenaunay in 1900.The origin of this syndrome continues to be investigated and many theories are discussed. Some authors believe that these venous abnormalities result from a deep venous obstruction or yet from deep venous atresia, leading to edema and hypertrophy of the extremity [5, 6]. 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 [7]. 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. Lesions follow a mosaic pattern, where heterozygotes of a single defective gene would be phenotipically normal, but the defective allele could be transmitted for many generations. The trait would only be expressed when a somatic mutation occurred in the normal allele, in the early embryogenic phase, originating a population of clonal cells for the mutation of KTS [8].

This syndrome occurs with little frequency in and gene theory suggests that a single gene with a lethal defect in homozygous individuals is involved in the case. In a study series of 252 patients at the Mayo Clinic, 63% of patients had all 3 features and 37% had 2 of the 3 features. Port-wine stain was seen in 98% of patients, varicosities or venous malformations in 72%, and limb hypertrophy in 67%[9]. Atypical veins, including lateral veins and persistent sciatic vein, were present in 72% of patients. Finally, deep venous abnormalities included aneurysmal dilation, hypoplasia, aplasia, and absent or incompetent valves [5].

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 nonsyndromic capillary malformations of the skin [11].

There is no definitive treatment for this disorder and patient's condition and 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. The superficial treatment of hemangiomas [12] for varicose veins, compression stockings are recommended for venous insufficiency. Surgical treatment is 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.

However here we report 2 cases of Klippel-Trenaunay Syndrome with common features port wine stains, limb hypertrophy and varicose veins with lymphangioma as an additional feature in former patient.

CONCLUSION

Patients with KTS should be monitored at least once in a year and more often if clinically indicated and desired. OPD basis follow-up of stable cases can be done. If the disease progresses, imaging studies should be performed and medical or surgical intervention should be pursued as per the demand. Moreover, researches in genetics should be encouraged so that in the future we may be able to understand the etiology of this disease.

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