Case Report

Klipple-Trenaunay Weber Syndrome, A Rare Disease with Literature Review

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Abstract

Klippel-Trenaunay Syndrome (KTS) is an uncommon spontaneous disease characterized by the clinical trial of capillary malformation, soft tissue and bone overgrowth, and atypical varicosities1. Although this was described almost a century ago, the exact incidence has not yet been estimated. Its pathogenesis has been described by a number of concepts. Clinical manifestations of this disease might range from a moderate, asymptomatic illness to potentially lethal bleeding and embolism. The management of this illness includes careful diagnosis, prevention, and treatment of complications associated with the disease2. Here we present an adolescent female 14 years of age who presented with right lower limb(around knee joint) swelling for the last 4 years which was progressively increasing in size when compared with the rest of the limbs.

Keywords: Klippel-Trenaunay Syndrome (KTS), Rare Disease

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Introduction

Klippel Trenaunay Weber Syndrome, a congenital disorder which is rare, is characterized by lower limb hypertrophy along with vascular anomaly and capillary malformations known as port wine stain. Some other anomalies like lymphatic block-adge and lipodermatosclerosis are often associated with it as well. It also includes varying degrees of vascular malformations in different systems inclu-ding gastrointestinal system, genitourinary and cent-ral nervous system[3]. This condition presents at birth but due to complex vascular involvement it is not only difficult to classify but also to diagnose it as well. KTS is a disease with wide range of capillary, venous, soft tissue and bony abnormalities. At the same time it is a disease with greater chance of misdiagnosis.

It is a sporadic disease with unknown etiology. Among various theories, Servelle's theory of venous system obstruction resulting in venous hypertension and therefore development of abnormal venous pathway and tissue overgrowth [4]. Berry et al comm-ented in 1998 that this phenomenon is due to alter-ation in vascular remodeling might be at the level of angiopoiten 2 antagonism[5].Already various case rep-orts are available in world literature but still incid-ence and genetic predisposition of this disease needs to be determined[6,7].

Case Report

A 14 year old girl who presented with right leg swelling and pigmentation for last few years which was gradually increasing. Her mother who is a house maid by profession took her to a disability manage-ment center where she was diagnosed to have this disease. She was suspected to have varicose veins along with vascular manifestations therefore she was referred to radiology center for Doppler scan lower limbs. She was advised X rays and Doppler studies of lower limb and further advised to continue conser-vative management for it. The diagnosis of typical KTWS was made on the basis of clinical and radiological findings which included the following: Port wine stains on skin of both hands and lower limb leg length discrepancy and swelling of right knee joint was noted. The circumferential measure-ments of the lower limbs were taken at different bony points which varied significantly. For example leg circumference was 7 cm more on right as compared to left side. Similarly thigh circumference was 2.5 cm more on right side as compared to the left side. More-over, vascular malformation in lower limb were more prominent around knee joint. The mid upper arm and mid-forearm circumferences were 18.5 cm and 18 cm, respectively, and showed no major variation. However, it was not associated with any limb length difference.

Doppler scan of the leg to establish patency, incompetence, thrombosis, arteriovenous shunting and any anomalies such as hypoplasia was done. Plain X-rays of the long bones in lower limbs was done. (scanogram).T2-weighted MR images showed malformed venous and lymphatic lesions as areas of high signal intensity. MR imaging also depicted deep extension of lowflow vascular malformations into muscular compartments and the pelvis and their relationship to adjacent organs as well as bone and soft tissue hypertroph.

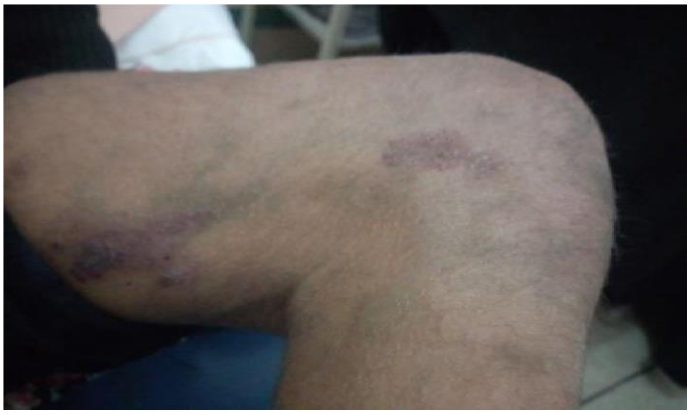

Figure 1: *Lower limb with KTS syndrome, Right knee varicosities and hemangiomas*

The best care for such patient is provided by a multidisciplinary approach-a pediatrician, a cardiologist ,a vascular and an orthopedic surgeon. In patients with concomitant lymphoedema, recurrent bouts of cellulitis and lymphangitis are common which need special care in prevention phase.

Surgery is only performed on KTS patients who exhibit symptoms and are not candidates for less invasive therapy. Prior to vascular operations, the deep system's patency and the degree of abnormallities must be carefully assessed. Facial asymmetry and hemangiomas in the central nervous system might make intubation difficult and increase the risk of complications by causing intraoperative bleeding. The best course of treatment for patients with normal deep veins is to completely remove the marginal vein surgically. Adequate vein exposure is advised, particularly for large perforators that need to be ligated, as this vein may have very large perforators to the deep veins.

Because deep veins can spontaneously expand to nearly normal size following resection, marginal veins can be removed if they are hypoplastic[9]. However, aplastic deep veins are a strict no-go for marginal vein excision.

For limb disparities of 1.5 cm or less, heel implants are usually enough in terms of limb hypertrophy. Orthopedic surgery might be recommended for larger disparities. Total knee arthroplasty has demonstrated promising outcomes for severe arthritis with overgrowth of one limb. However, epiphysiodesis(partial or complete) may be considered in cases where physis are still open. When the size of the damaged limb interferes with daily activities, it may be necessary to amputate it. Urogenital and gastrointestinal involvement is not as minimal as originally believed. According to recent research, urinary involvement is approximately 30% and gastrointestinal involvement can reach 20%. Endoscopic cauterization is typically necessary for these individuals, however occasionally refractory bleeding may call for emergency intervention.

Discussion:

A rare congenital disorder known as KTWS syndrome is characterized by faulty blood and lymphatic vessels. Its primary three characteristics include hypertrophy of various leg bones and soft tissues, port wine stain, which is brought on by capillary abnormalities that give the skin a reddish or purple hue, and various vascular anomalies. Maurice Klipple and Paul Trenaunay, two French physicians, were the first to record it in 1900. They documented two such patients who had identical triads of patients[9] A few years later, Parks Weber included AV malformation in this trio as well.

KTWS can be classified as either typical or atypical. Port wine stain is always included in conventional KTWS, however it is not in unusual types. The latter is quite uncommon. The condition has no racial or gender propensity. A diagnosis is usually made during pregnancy or during delivery. USG, Doppler investigations, CT scans, arteriography, and MR angiographic examinations that reveal common vascular abnormalities are used to make the diagnosis. The management is primarily conservative with compression garments Both lymphedema and chronic venous insufficiency respond well to it; however, lifestyle changes, appropriate wound care, dressings, and specific orthopaedic footwear may also be required.

Laser therapy for portwine stain, 100% alcohol (serial) sclerotherapy, foam sclerotherapy with sodium tetradecyle, and polidocanol are less invasive options for treating superficial venous ulcers. Antibiotics, corticosteroids, elevation, and analgesics can all be used to treat cellulitis and thrombophlebitis. We managed this case with conservative treatment initially in the form of POP casting followed by Hinged knee Brace for improving Range of motion coupled by passive physiotherapy. Patient was also monitored with regular Doppler scans to rule out possible development of vascular malformations.

Conclusion:

Klipple Trenaunay syndrome is a congenital vascular malformation disorder which can be managed effectively by combination of medications, surgery, laser therapy and physical and occupational management.

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Authors' contribution:

AR: Concept & design, conducted literature review, drafted initial document, created image, and amended the final draft

HMKS, KNC: Drafting of draft.

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