Klippel-Trenaunay-Weber Syndrome – A Rare Case with Visceral Manifestation and Thrombosed Marginal Vein of Servelle

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ABSTRACT

Klippel-Trenaunay-Weber syndrome is a rare congenital syndrome characterized by capillary, venous or lymphatic malformation and hemihypertrophy. Only 1000 cases of this syndrome have been reported in the literature. A rare case of this syndrome is presented here, who presented with its visceral and vascular complications and was diagnosed for the first time.

Keywords: Hemihypertrophy, Klippel-trenaunay-weber syndrome, Marginal vein of servelle, Port-wine stain.

How to Cite This Article: *Ibrahim MI, Awan HL, Khan B, Javed K, Zehra SM, Awan MH. Klippel-Trenaunay-Weber Syndrome – A Rare Case with Visceral Manifestation and Thrombosed Marginal Vein of Servelle. Pak Armed Forces Med J 2022; 72(3): 1142-1143. DOI: https://doi.org/ 10.51253/pafmj.v72i3.6694*

INTRODUCTION

Klippel-Trenaunay-Weber syndrome, also known as the angio-osteohy-pertrophy syndrome, is a rare congenital disorder affecting 1 in 100,000 population worldwide.[1] It usually affects a single extremity, most commonly the lower limb (75% cases); however, involvement of more limbs, trunk and face has also been reported.[2]

CASE REPORT

A 30-year-old male presented to CMH Rawalpindi with complaints of painful knee movements and could not bear weight on his left leg. In addition, the patient had a large port-wine stain along the antero-lateral aspect of the left lower thigh (Figure-1), atypical varicosities and hypertrophy of the left lower limb. The patient had a previous history of recurrent episodes of hematuria in his childhood, for which cystoscopy was also performed at the Sindh Institute of Urology and Transplantation.

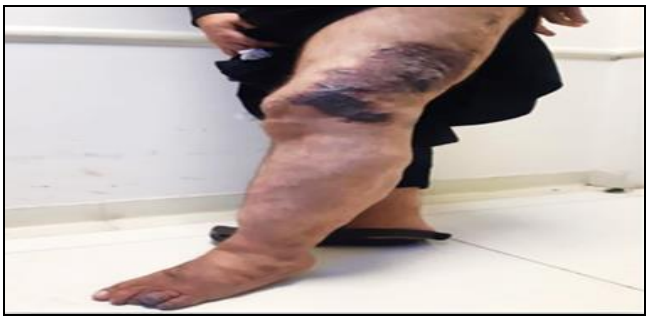

Figure-1: A large port wine stain on lateral aspect of left thigh with visible varicosities.

Computed tomographic arteriography (CTA) and Computed tomographic venography (CTV) of the lower limbs showed a leg length discrepancy of approximately 2 cm longer than its right side (Figure-2).

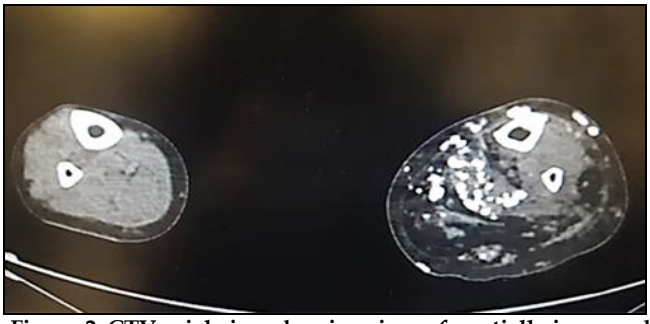

Figure-2: CTV axial view showing circumferentially increased soft tissue thickness of left lower limb with contrast opacified superficial varicosities and intramuscular AV malformations.

In addition, multiple dilated varicosities were noted in the atypical distribution along the dorsum of the foot, and lateral calf extending up to the left groin, left buttock and left half of the scrotum, with few of the vascular channels extending into the muscles of the leg along with small calcified phleboliths also seen (Figure-2). The most striking finding was the persistent sciatic vein (Figure-3) and marginal vein of Servelle (Figure-4).

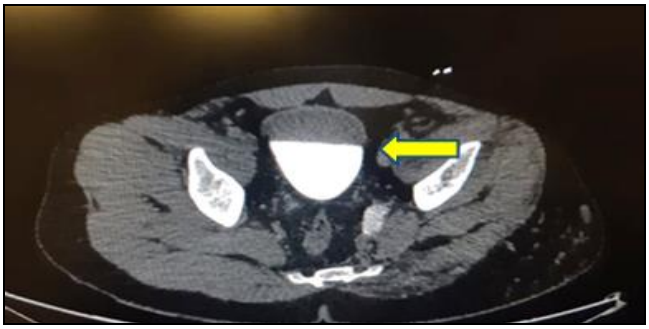

Figure-3: CTV axial view showing contrast opacified persistent left Sciatic vein (arrow).

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Received: 04 May 2021; revision received: 08 Jul 2021; accepted: 14 Jul 2021

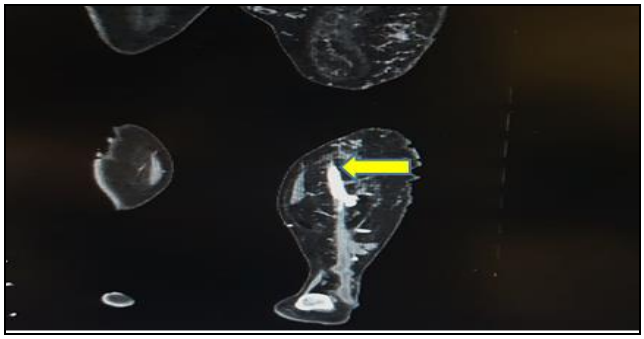

Figure-4: CTV coronal view showing thrombosis of the persistent lateral marginal vein (marginal vein of Servelle) along lateral aspect of left calf.

Klippel-Trenaunay-Weber syndrome was diagnosed with the typical triad of hemihypertrophy, a typical varicosities and capillary malformation (port-wine stain). In addition, Doppler ultrasound showed thrombosis due to thrombophlebitis of the lateral marginal vein, a known complication.

DISCUSSION

Klippel-Trenaunay-Weber syndrome is characterized by a typical triad of capillary malformation (port-wine stain), venous or lymphatic malformation and soft tissue and bone hypertrophy.[3] In 1900, French physicians, Klippel and Trenaunay, reported the first case of this syndrome.[4] Later in 1918, Weber described its association with arteriovenous malformations.[5] Klippel-Trenaunay-Weber syndrome affects 1 in 100, 000 population worldwide and has no gender or ethnic predilection and is assumed to be due to somatic mutation in the PIK3CA gene.[6]

Capillary malformations are the most common association and manifest as the port-wine stain. Venous malformations are a constant feature of this syndrome. In the superficial venous system, abnormalities include venous ectasia, persistent embryonic veins, i.e., marginal vein of Servelle and Sciatic vein, and significant venous malformations. Aneurysms, aplasia, hypoplasia, duplications, and venous incompetence may occur in the deep system. The marginal vein of Servelle courses over the dorsum foot and along the lateral aspect of the calf region and is considered pathog-nomonic of this syndrome. Lymphatic malformations, when present, resulting in oedema, increased soft tissue thickness and a predisposition to the development of cellulitis.[7] Visceral manifestations are rare, affecting only 1% of the cases and may involve the genitourinary system or the gastrointestinal tract.[8]

Complications include thrombophlebitis, thromboembolism, high output cardiac failure due to AV malformations and Kasabach-Merritt syndrome.[9]

Treatment options include regular compression stockings and graded compression of the affected limb, stripping and sclerotherapy of varicosities and laser ablation for port wine stains.[10]

Conflict of Interest: None.

Author's Contribution

MII:, LHA:, BK:, KJ:, SMZ:, MHA: Direct contribution.

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