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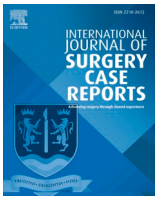

Case report

Klippel Trenaunay syndrome in a 3-year-old: A case report

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ABSTRACT

Introduction and importance: Klippel-Trenaunay syndrome (KTS) is characterized by a triad of port-wine stain, varicose veins and soft tissue or bony hypertrophy of lower limb. Varicose veins in Klippel Trenaunay syndrome are mostly distributed in the lateral aspect of the lower limb. The exact etiology of KTS is not known, and the treatment usually starts with conservative management- limb elevation, compression stockings and physiotherapy. However, some cases are severe enough to warrant surgical management.

Case presentation: Here we present a case of a 3-year-old male child with clinical features suggestive of Klippel Trenaunay Syndrome managed successfully with sclerotherapy of persistent lateral marginal vein of servelle. At one month follow-up the vein was sclerosed and there was a significant reduction in varicosities of leg.

Clinical discussion: Starting treatment of varicose veins in Klippel Trenaunay Syndrome in children is effective in preventing long-term complications in adults. Thus, treating venous malformation with sclerotherapy is warranted in early childhood to prevent venous hypertension and chronic venous insufficiency.

Conclusion: Varicose veins in KT syndrome can be managed successfully by sclerotherapy of lateral marginal vein of servelle resulting in significant reduction in varicosities of leg.

1. Introduction

Klippel-Trenaunay syndrome (KTS) is a rare sporadic disorder which is present from birth [1]. International society for the study of vascular Anomalies, 2018 has classified it under combined vascular malformations involving venous, capillary system and limb overgrowth with or without lymphatic involvement [2]. At least two of the above features should be present to clinically diagnose KTS [3]. It is mostly unilateral involving lower extremities and rarely can involve bilateral lower extremities, upper extremities, trunk and internal organs [4]. The etiology of KTS is not clearly known; however, mesodermal abnormality during fetal development is said to be the cause for maintenance of arteriovenous communication in limb bud [5]. Conservative treatment along with endovascular therapy is employed which focuses on relieving the symptoms and preventing the complications for improving the quality of life [1]. Here, we present a case of 3- year-old male child with bluish purple discolouration of right thigh and leg since birth with prominent dilated and tortuous cutaneous veins of the same region mostly in the lateral aspect which is managed successfully by two sessions of sclerotherapy. This article was written in accordance with the SCARE 2020 Guideline [6].

2. Presentation of case

A 3-year-old male child presented with bluish purple discoloration of right thigh and right leg since birth (Fig. 1). It was associated with prominent dilated and tortuous superficial veins of thigh and leg distributed mostly in the lateral aspect. There was no history of trauma, any surgeries in the legs or ulcerations. Arterial pulse in both limbs was of normal character and volume.

Doppler ultrasonography showed multiple dilated superficial veins. Presence of single dilated vein on the lateral aspect of right thigh and leg suggested persistence of lateral marginal vein of Servelle. Maximum diameter of this vein was 10 mm (Fig. 2). All the features were suggestive of Klippel Trenaunay Syndrome. There was no significant enlargement and tortuosity in great saphenous and short saphenous veins.

The varicosities especially in the lateral aspect of thigh and legs were treated by foam sclerotherapy by injecting polidocanol 3 % in the lateral marginal vein of servelle at multiple sites under USG guidance.

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Compression was applied at the injection site using a sterile gauze followed by application of compression bandage.

A total of two sessions of sclerotherapy was done over a period of one week. On follow up at 2 weeks, significant reduction in varicose veins in right thigh and leg was noted. USG showed reduction in diameter of the lateral marginal vein of servelle (5 mm) which was non compressible indicating successful sclerosis of the vein.

3. Discussion

Klippel-Trénaunay syndrome is a rare congenital disorder that was first reported in 1900 by Maurice Klippel and Paul Trenaunay [1]. It is characterized by the triad of port-wine stains, hypertrophy of soft tissue, and varicosities [1]. The incidence has been estimated at 2–5 per 100,000, more often in male with no racial predilection [1].

The exact etiology of KTS is still unknown however genetic study of cases of KTS have revealed that there is a gain of function somatic mutation in PIK3CA gene in a mosaic pattern causing excessive proliferation of vascular tissue leading to capillary, venous and lymphatic malformations involving only some part of the body [7,8]. While port-wine stain is present since birth, varicose veins may not be evident until child starts weight bearing and becomes aggravated only during adolescence reflecting hormonal influence over vascular proliferation [9].

There are no consensus diagnostic criteria for the management of the disease considering its unapparent etiology [1]. In most cases, diagnosis is made by clinical and radiological evaluation [10]. Imaging modalities such as ultrasound, MRI, and CT can reveal soft tissue hypertrophy, vascular malformations and associated complications like pulmonary embolism and deep vein thrombosis [10–12].

Additionally, there is no accepted protocol for treating these cases

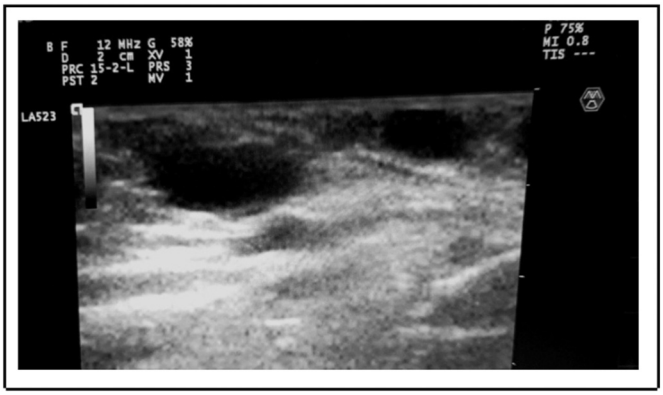

Fig. 2. Ultrasonography of right lower limb showing dilated lateral marginal vein of servelle with dilated accessory vein.

[1]. The relative indications for treating KTS include Pain, functional impairment, swelling, limb asymmetry and cosmetic defect whereas absolute indications are hemorrhage, infection and acute thromboembolism [1].

Treatment of KTS includes a combination of conservative and surgical approach [1,4].

Conservative management includes non-invasive measures such as the use of compression stocking, and intake of analgesics, antibiotics and corticosteroids [1,4]. Surgery is usually performed for symptomatic cases and is guided by preoperative evaluation of deep venous system using computed tomography (CT) arteriography [1,11]. Additionally, duplex scanning contrast geography can be done for assessment of

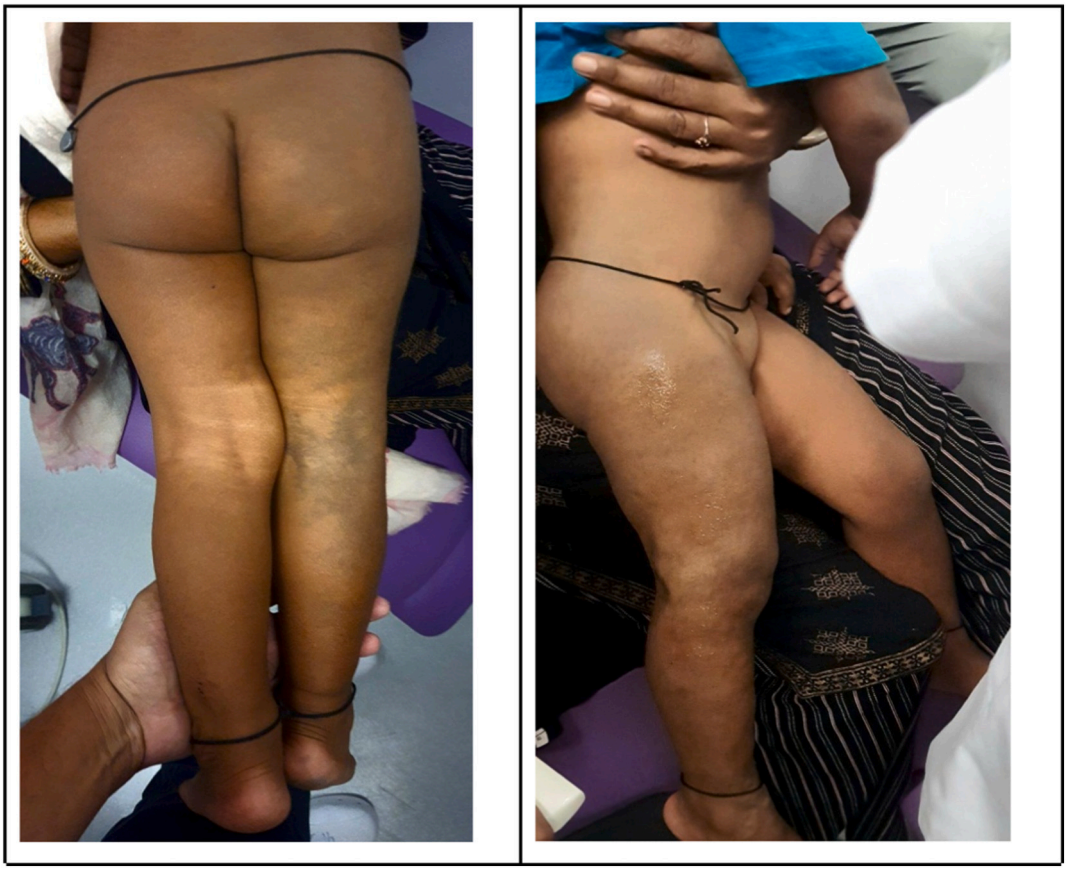

Fig. 1. Right lower limb showing bluish-purple discoloration of skin with prominent dilated veins along the right lateral aspect. (For interpretation of the references to colour in this figure legend, the reader is referred to the web version of this article.)

extent of vascular involvement and presence of arteriovenous fistula [1,11]. Ligation and stripping has fallen out of favor because of worsening of symptom following these surgeries [13]. Laser therapy can be employed for the treatment of port wine stain and ulceration but not for lesion under the skin [1]. Endovenous laser therapy for the varicosity, radiotherapy for the hemangioma are of great use but the result are slow [1].

There have been studies of this syndrome in Nepal [4], however KTS in a child as young as 3 years treated with sclerotherapy has rarely been reported. Literature suggests that endovenous laser ablation adjunct with sclerotherapy is an effective management of KTS in pediatric patients [4,10]. However, in our case the lateral marginal vein was tortuous which could have created trouble in insertion of radiofrequency ablation (RFA) catheter. Thus, we opted for doing sclerotherapy as the mainstay of treatment to correct underlying varicosities.

4. Conclusion

Klippel Trenaunay Syndrome is a rare vascular malformation which is usually under-evaluated in pediatric patients. Diagnosis in early life requires a high index of suspicion combined with detailed clinical and radiological examination. Additionally, varicosities involving lateral marginal vein of servelle in KTS can be managed in early childhood with USG guided sclerotherapy injection yielding good outcome.

Consent

Written informed consent was taken from the patient and none of the identifying characteristics were included.

Ethical approval

N/A.

Funding

None.

Author contribution

Robin Man Karmacharya: Surgical procedure, patient care, manuscript writing,
Satish Vaidya: Surgical procedure, patient care, manuscript writing,
Binay Yadav: Surgical procedure, patient care, manuscript writing and editing
Sahasra Joshi: Manuscript editing
Diwas Karkee: Manuscript editing
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Swechha Bhatt: Manuscript writing and editing

Guarantor

Dr. Robin Man Karmacharya.

Research registration

N/A.

Provenance and peer review

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Declaration of competing interest

None.

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