

Case Report

A rare case of Klippel-Trenaunay syndrome with thrombosed venous mass

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ABSTRACT

Klippel-Trenaunay syndrome is characterized by a triad of port-wine stain, varicose veins with or without venous malformations, and bony and soft tissue hypertrophy. It usually affects only one extremity. Its cause continues to be investigated, although theories abound. This disease has several morbidities like bleeding, venous thrombosis, and in some cases enlargement of limb that may require amputation. We report a case of 24 yrs old male patient with KT syndrome involving the left lower limb with thrombosed venous malformation.

Keywords: Varicose veins, Venous malformation, Foam sclerotherapy, Radiofrequency ablation

INTRODUCTION

Klippel-Trenaunay syndrome (KTS) is characterized by triad of port-wine stain, varicose veins, and bony and soft tissue hypertrophy. Herein we report a case of KTS with a rare presentation.

CASE REPORT

A 24 years old male patient presented with recent onset of Pain, firm mass and recurrent bleeding from the left leg since 1 month and dilated veins with enlargement of the left lower limb for 10 yrs. On examination he had dilated lateral sciatic vein from upper thigh extending up to left ankle with intervening thrombosed venous mass over the left lateral aspect of upper calf with port vein pigmentation (Figure 1) and lengthening of the involved limb of about 2 cm. CT Venogram showed enlarged lateral sciatic vein joining deep femoral vein with thrombosed venous mass at upper calf and hypoplasia of

the deep veins, with multiple huge perforators incompetence (Figure 2).

Operative details

Patient was taken up for endovenous radio frequency ablation of lateral sciatic vein using 2 sheaths of 5F size with 100cm continuous RF ablator with foam sclerotherapy, using 3% sodium Tetra decyl sulphate and excision of thrombosed venous mass (Figure 3 & 4) under regional anesthesia. Patient had uneventful postoperative recovery (Figure 5 & 6) with disappearance of pain & swelling and post-operative CT venogram showed obliteration of lateral sciatic vein.

DISCUSSION

Klippel-Trenaunay syndrome (KTS) is characterized by a triad of port-wine stain, varicose veins, and bony and soft tissue hypertrophy. It generally affects only one extremity. Lesions are present at birth and in

approximately 75% of patients symptoms starts before 10 years of age. The difference between KTS and Klippel-Trenaunay-Weber syndrome (KTWS) is that the latter includes significant arteriovenous malformations in the affected extremity.



Figure 1: Pre operative photo showing Thrombosed mass with varicosities.

Klippel-Trenaunay syndrome is a rare disease, with estimates of approximately 1 per 30,000 live births. The disease has a classic triad of capillary malformations, vascular anomalies (classically varicosities such as the persistent embryonic lateral vein of Servelle), and hypertrophy of bony and soft tissues. Patients with Parkes-Weber syndrome have similar presentations as those with KTS and can often be indistinguishable on physical examination. However, advanced imaging has made the differentiation between high- and low-flow arteriovenous malformations (AVMs)—the distinguishing feature—much easier. Low-flow AVMs are seen in KTS and have relatively low morbidity, whereas high-flow AVMs are more appropriately assigned as Parkes-Weber syndrome. Differentiation is important, as high-flow AVMs can cause more serious clinical consequences such as high-output heart failure, more prominent skin manifestations with an increased chance of skin ulcerations, and increased limb-length discrepancies.

The expression and presentation in KTS have a widely varying disease spectrum, from incidental to incapacitating. Symptoms such as pain, skin breakdown,

and lymphedema can cause relatively little impairment. However, superficial thrombophlebitis with overlying cellulitis, deep venous thrombosis, and pulmonary embolism can be more severe requiring hospitalization. There are also associated congenital abnormalities, including developmental dysplasia of the hip and syndactyly, roughly 30% of the time. It is important that the physician is aware that vascular malformations can occur in internal organs and may be the source of major bleeds in anemic patients with KTS.



Figure 2: CT venogram of patient showing lateral sciatic vein.

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The etiology of the disease is still under investigation, but it is well accepted that venous abnormalities are not the causative insult. Embryonic mesodermal changes resulting in increased angiogenesis lead to increased vascular flow causing tissue hypertrophy and vascular



Figure 3: RF ablator through 5f sheath in sciatic vein.



Figure 5: Post operative CT venogram - no lateral vein seen.



Figure 4: Thrombosed venous mass being excised.



Figure 6: Post operative picture of patient.

changes. Most experts agree that the majority of cases of KTS are due to sporadic polygenic mutations. A theory of

paradominant inheritance seeks to explain KTS by the formation of a lethal mutation in a gene, whereby homozygous embryos do not survive but heterozygous embryos survive and are normal. It is not until a second hit with loss of heterozygosity occurs that phenotypic expression results. The paradominant theory helps explain why rare family links to the disease exist, as well as the mosaicism that can occur in individuals.

A single treatment plan is not recommended, as the specific type and severity of symptoms should guide management in KTS. Now a days endovenous ablation has gained popularity due to less post-operative pain and blood loss. For superficial varicosities, conservative measure (compression therapy) is tried prior to intervention and is performed only after radiographic confirmation of a patent deep venous system. Regarding limb-length discrepancies, a cutoff of 3.0-cm difference has been suggested for epiphysiodesis. Sclerotherapy can be used on capillary malformations for cosmetic reasons and adjuvant therapy to endovenous ablation. Surgical debulking often fails or worsens symptoms as venous and lymphatic channels are destroyed, leading to further swelling and poor wound healing. Overall, treatment is often not definitive and 50% of patients re-experience symptoms after surgery despite reported clinical and symptom severity improvement in many patients.

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